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**This thesis is in a form
acceptable for
the award of the degree.**

CARRIERS OF RESPONSIBILITY:

**An existential encounter with parents who know
their child is, or could be, a carrier of a mutation
in the cystic fibrosis gene**

by

MELANIE JANE ATTARD

Submitted in fulfilment of the degree of Doctor of Philosophy

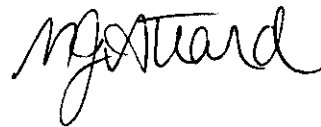
THE UNIVERSITY OF SYDNEY

2009

DECLARATION

I certify that this thesis has neither been previously submitted for a degree nor has it been submitted as part of the requirements for a degree.

I also certify that the thesis has been written by me. Any help that I have received in my research and work and the preparation of the thesis itself has been acknowledged. In addition, I certify that all information sources and literature used are indicated in the thesis.

A handwritten signature in black ink, appearing to read "M. J. Stuard". The signature is written in a cursive style with a large initial "M" and a long, sweeping underline.

DEDICATION

This work is dedicated to the memory of a very strong and very wise woman who has shaped my life enormously, Katarin Attard (1920 – 2007).

I loved spending time with you at your house “hdejn it-turretta” in Ghaxaq, Malta.

I was so proud to have you as my Nanna Kekina.

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I would firstly like to acknowledge the traditional owners of the land where I have lived for much of my life, the Kurna people, and also the Cadigal and Eora people, upon whose land The University of Sydney now sits, and where I completed my studies towards this PhD. May the dream of true Reconciliation in this country one day be a reality that brings healing for us all.

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To the person who has been a special part of who I am for over thirty years – my sister, Leanne, just because. And lastly to the two little ones, Luke and Anna, who will always bring joy into our lives.

And here are trees and I know their gnarled surface, water and I feel its taste. These scents of grass and stars at night, certain evenings when the heart relaxes—how shall I negate this world whose power and strength I feel? Yet all the knowledge on earth will give me nothing to assure me that this world is mine. You describe it to me and you teach me to classify it. You enumerate its laws and in my thirst for knowledge I admit that they are true. You take apart its mechanisms and my hope increases. At the final stage you teach me that this wondrous and multicoloured universe can be reduced to the atom and that the atom itself can be reduced to the electron. All this is good and I wait for you to continue. But you tell me of an invisible planetary system in which electrons gravitate around a nucleus. You explain this world to me with an image. I realize then that you have been reduced to poetry: I shall never know. Have I the time to become indignant? You have already changed theories. So that science that was to teach me everything ends up in a hypothesis, that lucidity founders in metaphor, that uncertainty is resolved in a work of art. What need had I of so many efforts? The soft lines of these hills and the hand of evening on this troubled heart teach me much more. I have returned to my beginning. I realize that if through science I can seize phenomena and enumerate them, I cannot, for all that, apprehend the world. Were I to trace its entire relief with my finger, I should not know any more. And you give me the choice between a description that is sure but that teaches me nothing and hypotheses that claim to teach me but that are not sure. (Camus, 2004a, p.453-454)

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ABSTRACT

My aim in this study was to interpret how the individual's experience of being-a-carrier and parenting-a-carrier is shaped by what it means to be human – to look for the existential categories of experience which pervade the hopes and concerns of parents within 'carrier families'. In order to do this, I devised a method of understanding based on concepts that I explicated from my reading of the existentialist literature. Eighteen parents/grandparents participated in the study, with thirteen open-ended, in-depth interviews conducted in total. The emerging data was found to be replete with the dilemmas and paradoxes of human existence and pointed wholeheartedly to existentialist philosophy as a fitting explanatory device. In turn, the data itself validated and became illustrative of the concepts I had delineated. The existential concepts of *Despair*, *Self-estrangement* and *Authenticity*, *Bad Faith* and the *Predominance of the Other*, *The Look*, *Fear* and *Anguish (Angst)*, *Nausea*, *Absurdity* and *Revolt*, *Ambiguity*, *Freedom* and *Responsibility/Choice*, *Care* and the *Technological Attitude* were all examined in relation to, and as they defined, the topics uncovered and discussed in the interviews with the participants. I used the existentialist 'ethic' and the notion of a child's 'right to an open future' as a means to examine the attitudes and actions of parents and understand when these work to expand or to limit the 'possibilities' of their children. The findings of the study provide an insight into what is happening in 'carrier families' when genetic information becomes known, consider the ways that people attempt to transcend their genetic situation, examine how people look to Technology in order to evade genetic threat, and consider the limitations of Technology in countering that threat. I examine the existentialist notion of the Other and its influence on the individual's experience of the carrier state and choices they make in relation to it. I draw parallels between existing *authentically* in the existential sense and achieving autonomy in the genetic counselling/testing context; both so difficult to achieve from a position of immersion in the 'they'. I trouble the 'promise' of Technology and show that parents are 'carriers of responsibility' with respect to their child's genetic information, with an obligation to

care for it and their child's right to an open future accordingly. I explain that the 'choices' ahead of the children we know as 'CF carriers' will not necessarily be ones made from a situation of *Freedom*, given that the processes of geneticisation and technologisation taking place about them are as likely to work to restrict their possibilities as to maximise them. In this vein, I show that genetics serves to 'produce' identity rather than to 'reveal' it; that it creates the difference between carriers and non-carriers rather than uncovers it. I propose application of the existentialist ethic in the genetic counselling/testing context, with the hope that *Existential Freedom* and the child's right to an open future can be used to guide parents' decisions and professionals' practice in that arena. I emphasise that this problem is about much more than genetics and that therefore a solely geneticised response cannot be the solution.

PREFACE

I am a geneticist who has used the philosophical framework provided by existential thought to interpret the experiences of parents who know their child is, or could be, a carrier of a mutation in the cystic fibrosis gene. The broad aim of this thesis is to make an ethical and knowledge-based contribution to the debate about the genetic testing of children and to provide health professionals and genetic counsellors with an insight into the experiences of 'carrier families'. In this thesis, I have also developed a conceptual framework for analysing qualitative data by explicating a set of concepts from the existentialist literature. In addition, I have delineated a new health care ethic – 'the existentialist ethic' – for use in the genetic counselling/testing context, the crux of which is readily understood, accessible and practicable.

All work that is not my own has been referenced in the relevant section of the text and all sources have been listed in the Reference List at the end of the thesis. Ethics approval was obtained from the relevant hospital and university ethics committees prior to conducting the research. I have been financially supported by the Australian Government in this undertaking, having been the recipient of an Australian Postgraduate Award. Some of the work undertaken in relation to this thesis was presented at a conference, and the abstract published in the Conference Proceedings:

Attard, M. (2001). *What do we mean when we label a person as a carrier of a genetic illness? Contesting Conversations in Practice, Education, Research and Policy (Nursing & Midwifery): Proceedings of the Fourth International Conference, Adelaide, South Australia.*

I will also be presenting some of the findings from this study in April 2009 at the 3rd International "In Sickness & In Health" Conference to be held in Victoria, British Columbia, Canada. The abstract submitted was entitled *How is the experience of parents who know their child's carrier status for cystic fibrosis shaped by what it means to be human?*

A WORD ON OPPRESSION

The existentialist authors were products of their own time, constrained by the temporal limitations of their own existence. They were predominantly men and predominantly indifferent to the oppression of women, if not blind to it (although Simone de Beauvoir eventually brought the 'secondary' nature of women's existence to the foreground of existentialist thought in *La Deuxieme Sexe*). Their now-archaic use of the pronoun 'man', rather than 'human beings', and 'mankind', rather than 'humanity', appears many times in quotations in this text. I concur with Crotty's words on this and have chosen to treat this matter here in the way that he has done previously:

Oppression takes many forms and is incarnate in the very language we speak and write. As far as the oppression of women is concerned, a number of authors quoted in these pages wrote at a time when there was little, if any, consciousness of this. They blithely talk of 'man' when they mean women and men, and feel free to use the generic masculine whenever they need pronouns. My readers will readily recognise such usages, and no doubt deplore them, as they occur in the quotations given. There is hardly need for me to interrupt the text *ad nauseam* by inserting '(sic)' many times over to point them out (Crotty, 1996, preface).

Another form of oppression that may be seen to be incarnate in this thesis is the hetero-normative assumption (and the 'would-be-grandparent'-normative assumption!) that all children will one day become adults who wish to find a partner of the opposite sex who they can 'make babies' with. For the sake of elegance, it was not desirable to qualify each occasion in the thesis where this hetero-normative ideal could be seen to be lurking but I will say now that the one-in-100 chance that your average CF carrier has, *à priori*, of having a child with CF themselves, relies on a number of assumptions: that all adults wish to find a sexual partner; that that partner will be of the opposite sex and also Caucasian (therefore having a much higher chance of carrying a CF mutation than if they were from an alternate ethnic background); and that those people, as a couple, will wish to have a child together. Even contemplating whether or not a person who is a CF carrier will want their

partner to have a genetic test or not presumes that all people wish to be in a stable, monogamous relationship when they conceive a child. Suffice to say, it is not my intention that this thesis be seen as passively homophobic or racist although I think in some ways it acknowledges my suspicion that many, many Caucasian parents in Australia today do still expect that their children will grow up to be heterosexual and to have a Caucasian partner.

A WORD ON REFERENCING AND STYLISTIC MATTERS

When quotes from the participants in this study are included in the analysis chapters, the reference to the page number and line numbers in the corresponding interview transcript is shown in round brackets at the end of each quote. For instance, if a statement is attributed to Anne and referenced as '(15: 22-29)', this will mean that the quote can be found in lines 22 through to 29 on page 15 of the interview transcript with Anne & Kevin (biographies of the participants are provided in Chapter Four, which indicate whether a person was interviewed alone or with another).

All words and comments placed in square brackets in the interview transcripts were not actually spoken in the interview. Sometimes they refer to a non-verbal 'event' that may be of some import to the reader (i.e., they may allude to a pause, a laugh, the tone of voice or a gesture on the part of the speaker, or explain a temporary interruption to the interview). Alternatively, the square brackets may contain information that has been inserted in place of, or in addition to, spoken words where it would facilitate the reader's understanding of what was being said or to keep a reference to a particular family member (who was not assigned a pseudonym) or health professional anonymous.

In all quotes used throughout the entire thesis, whether from another author or from the interview transcripts, the same protocol is used to indicate when I have chosen to omit part of the original statement (for reasons of either clarity or brevity) midway through the citation. In these instances, '[...]' is used to show

where this has been done. Sometimes quotes that were used required a tense change so they would read correctly in the context in which they were placed in the thesis. On these occasions, the word that was changed has been placed in square brackets. At no time were any alterations to original quotes intended to change or embellish the meaning of the text in any way.

Bold text has always been used in this thesis for purposes of emphasis. Where such emphasis has been added to text within a quote from another author, I have indicated this at the end of the quote. *Italics* have been used for Latin terms and other non-English terms; for stylistic reasons for quotes that sit immediately under headings; as well as for the titles of longer works, such as books. Quotation marks have been used around the titles of shorter works, such as reports. For clarity and emphasis, *italics* have been used to denote reference to an existential concept. The word is capitalised where the concept is being referred to in its noun form (i.e., *Nausea*); otherwise it is not capitalised (i.e., *nauseous*). However, all references to the existential concepts in direct quotations of other works have been left in their original state. Similarly, Americanised versions of words in direct quotations have not been altered and '(sic)' has not been inserted in these instances. Indeed, all original content of direct quotations (e.g., spelling, emphases and quotation marks) has been exactly replicated here, unless indicated otherwise or in the circumstance where double quotation marks in the original have been changed to single quotation marks here.

CHAPTER ONE: CONTEXT AND SIGNIFICANCE

'GENETICS' AND 'GENETICISATION'

In the 1990s, the word 'genetics' came to be associated with images of sterile, soul-less human clones for whom Dolly was an ovine prototype, or otherwise, genetically-modified organisms set to render their primitive Darwinian-evolved counterparts redundant while providing a means for multinational corporations to control primary industry all over the world. Against this backdrop, it is little wonder if, at times, the work of geneticists is viewed somewhat dubiously.

In 1991, Abby Lippman noted that the way we talk about health and disease is increasingly "in the language of genetics" and that this is conditioning the way we think and behave (p.17-18). She said:

the dominant discourse describing the human condition is reductionist, emphasizing genetic determination. It promotes scientific control of the body, individualizes health problems and situates individuals increasingly according to their genes. Through this discourse, [...] clinical and research geneticists and their colleagues are conditioning how we view, name and propose to manage a whole host of disorders and disabilities. [...] "[G]enetics" is increasingly identified as *the* way to reveal and explain health and disease, normality and abnormality. (1991, p.18)

Lippman believes this conditioning "directs how intellectual and financial resources are applied to resolve health problems" and also "profoundly influences our values and attitudes" (1991, p.18). It was Lippman who introduced the term "geneticization"¹ to describe the ongoing processes by which "differences between individuals are reduced to their DNA codes" and "interventions employing genetic technologies are adopted to manage problems of health" (1991, p.19). Geneticisation also privileges genetics, at the expense of other narratives, as "the

¹ I will not use the Americanised spelling elsewhere in this thesis.

source of illumination” for understanding the “prevailing social concerns of our age” (Lippman, 1991, p.19).

The age of geneticisation coincides with an era of professional accountability and it is imperative that behind the production and utilisation of new genetic knowledge there lies reasoning which is ethically sound. It is a pertinent time for geneticists to take responsibility for their collective reputation in the community and reflect on how they themselves have been conditioned, on the assumptions and motivations that underlie their approach to research, and on the impact their work has on society. This thesis is a part of that action.

GENETIC TESTING

The rapid advances being made in health care since the advent of genetic testing will soon have a significant impact on every individual in our society. For many of us, the previously inaccessible and unknowable knowledge residing in our DNA will play a prominent role in shaping some of the most pivotal decisions we ever make in our lives. For some, it already has. Our lifestyle, our desired career path, our choice of partner, our resolve to have children, our reasons for prenatal testing, and our decision to continue with or to terminate a pregnancy may well be based on information derived from genetic testing.

Those who have already chartered this new, unknown, and seemingly infinite genetic territory have largely done so without lessons learned from previous generations to cushion them. For those who lived before us, to possess such knowledge and to be faced with such decisions was neither in their realm of experience nor hardly in their realm of contemplation. Yet even today, as the number of available genetic tests increases, and “[d]espite a century of popularisation and school education in Mendelian genetics” (Richards, 2000, p.2), public understanding of genetic inheritance and the far-reaching implications that genetic disease can have for an entire family remain limited. In our increasingly

genetic climate, it is important that ethical issues surrounding genetic testing can be debated by the entire community. Rosalyn Diprose says that

[t]hrough an increasingly vigorous and public debate about the ethics of genetics, we² have been asked to share in the geneticist's competence as well as in responsibility for the always uncertain consequences of scientific research (2005, p.244).

For this to occur, a sound 'genetic literacy' of the general public is required, as well as an empathic understanding of the varying circumstances in which an individual may consider taking a genetic test. This is why it is so important that we harness the experiences of those already affected by genetic testing, through listening to their stories and distilling meaning from them, **now**.

WHAT IS MEANT BY GENETIC TESTING?

Different working definitions of 'genetic testing' can be applied in different contexts. In the context of this research, the term 'genetic testing' is used in reference to analysis of the genetic constitution of the individual for purposes related to health care and reproductive decisions only. It will exclude circumstances where DNA 'fingerprinting' is used for paternity or forensic testing, or where polymorphisms in human proteins or DNA are studied in the context of anthropological or population genetics. Also, since in terms of ethical and social issues raised, "[t]he use of molecular genetic methods to identify microbial infections or genetic changes confined to tumours [are] not so different from the conventional investigations carried out in any pathology laboratory" (Clarke, 1998, p.1), these will also be excluded from the definition of 'genetic testing' used here.

For the purposes of discussing human inherited disorders and the medical, ethical, social and legal implications which evolve with the technology to identify people who carry specific disease-related genes, **a useful definition of genetic testing** has already been provided:

² It is unclear who Diprose refers to here but I have taken it to mean 'we', the general public.

Genetic testing is the analysis of a specific gene, its product or function, or other DNA and chromosome analysis, to detect or exclude an alteration likely to be associated with a genetic disorder (Harper, 1997e, p.8).

However, the situations in which genetic tests of this sort are performed do differ markedly. An individual may seek testing because of their own specific personal and family circumstances, or one may be part of a population-based screening program targeted at groups such as newborn infants or pregnant women where diagnosis of genetic conditions may be useful.

Genetic tests fall broadly into a number of categories: diagnostic³, presymptomatic⁴, predictive⁵, and carrier testing. Diagnostic testing provides answers to questions posed in the present tense about an individual's state of health. In contrast, presymptomatic and predictive genetic testing have the potential to forecast an individual's future health status. The difference between the two is the certainty with which the future is seen. The other type of genetic test is one that identifies a healthy individual's **carrier status** with respect to a genetic abnormality that will not affect their own health but could jeopardise the health of their own children. Carrier testing for autosomal recessive diseases, X-linked recessive diseases (in females), and balanced familial chromosomal rearrangements is usually performed when an individual believes the result will be relevant to their

³ A **diagnostic** genetic test is usually performed in order to confirm a clinician's suspicions that a patient is manifesting symptoms of a particular disorder. The test can only be considered diagnostic if the patient already has clinical symptoms or signs related to the disease which they have been tested for (Harper, 1997e, p.9). An exception to this, perhaps, is in the prenatal and neonatal diagnostic contexts of genetic testing, when there may not yet be overt features of the disease present. In these instances, genetic tests may have been initiated due to: a family history of a particular disorder; an abnormal maternal serum screen during pregnancy; or a routine neonatal screening test. These tests probably fall into the diagnostic category if symptoms of the disorder in question usually begin to manifest in the perinatal period.

⁴ A positive result in a **presymptomatic** test reveals just that – that an individual, although currently healthy, carries a genetic abnormality which almost certainly guarantees the development of disease symptoms at some point in their life (Harper, 1997e, p.9). The question with monogenic disorders such as Huntington's disease is not **if**, but **when**.

⁵ **Predictive** testing is less definitive than presymptomatic testing, operating in probabilities rather than certainties (Harper, 1997e, p.10). A result may indicate that an individual has a genetic predisposition to a multifactorial disease, one which is influenced by a number of genetic and environmental factors, and hence cannot be predicted accurately on the basis of genetic testing alone, if at all. It allows an estimation to be made of one's life-time risk of developing a particular disease, and/or one's relative risk compared with certain family members or populations.

own or to other family members' future reproductive decisions. The stakes are higher for those who are carrier tested for an X-linked recessive disease or chromosomal rearrangement because a positive result will mean they have a very high chance of conceiving an affected child regardless of their partner's genotype. Those who carry an autosomal recessive disease gene are healthy themselves and only at risk of conceiving an affected child if their partner is also a carrier (or sufferer) of the same disease. Most families do not even realise a particular recessive disease gene is scattered through a branch of their family tree until an affected child is born. Thus, the fateful meeting of two loss-of-function mutations at the same genetic locus in one individual reveals a latent reproductive threat that has cascading repercussions through both sides of the family.

WHAT MAKES GENETIC TESTING SO DIFFERENT TO OTHER FORMS OF MEDICAL TESTING?

There are a number of features which distinguish genetic testing from other forms of medical tests. It is these distinctions which give genetic testing special ethical and psychosocial considerations of its own. Common to all the types of genetic tests outlined above is the profound implications they are likely to have for family members other than the individual tested. While we have always made guesses at our various disease risks based on observations of what tends to 'run in the family' (life insurers do it all the time), genetic testing can provide levels of certainty and precision we never had access to before. Genetic tests may define exactly what the familial genetic defect is; they allow genetic counsellors to frame risk assessments in terms of an individual's genetic constitution rather than their position in a pedigree⁶ alone. However, it is conceivable that testing could disturb the way that genetic burden is shared across a family structure. For while the cloud of genetic disease may be lifted from some family members, it will almost inevitably rain down more heavily upon others; the climatic shift potentially dichotomising the family unit.

⁶ Family tree.

Another feature of genetic analyses that distinguishes them from more conventional medical investigations is that they can be carried out at any stage of an individual's life (although diagnostic genetic tests, by their very definition, are performed only at a time when symptoms or signs of illness are evident, the same genotypic result would also be obtained if the individual was to be tested at any other point in their life) (Clarke, 1998, p.1). This means that genetic testing can be used in the prenatal period to identify children who will be born with a genetic condition, raising questions about the termination of pregnancies. It also means that a genetic test can reveal, sometimes years or decades in advance, whether an individual is destined to develop a particular disease, raising questions about how helpful or harmful it is to have such foreknowledge. For some conditions, early information about an individual's genotype can bring great gains if there are treatment, prophylactic, or lifestyle modification options available to delay or even prevent the onset of symptoms, and to minimise or stave off the severe sequelae of these. It is clear that knowledge of this sort has the potential to be highly beneficial in these circumstances. It is the reason why neonatal screening programs can bring substantial gains for infants born with a variety of genetic conditions, early diagnosis precipitating early intervention even before the distressing onset of symptoms.

However, as eloquently stated by Nancy Wexler (1989, ¶ 8), “[t]he most disquieting dilemma provoked by genetic advances is the capacity to prophesy before we can prevent”. Wexler has written a number of intriguing and illuminating essays (1979; 1989; 1992) on what she terms “this painful hiatus in science” (1989, ¶ 8); this phenomenon of human genome research whereby “[t]he acquisition of genetic knowledge is [...] outpacing the accumulation of therapeutic power – a condition that poses special difficulties for genetic knowing” (N. S. Wexler, 1992a, ¶ 1). It is critical, particularly now that the Human Genome Project has been completed, that the psychological and emotional impact of this special type of “genetic knowing” – of knowing one carries a gene for a fatal or debilitating condition which cannot be prevented or cured, whether it be autosomal or X-linked, dominant or recessive – is understood.

A healthy person at high risk of carrying a dominant disease gene must consider very carefully whether it will be more helpful or harmful to take a presymptomatic test if there is a good chance they will discover they are going to develop a disease for which there is no chance of prevention, little treatment and no cure. One could not have designed a better archetype of hereditary illness for studying the psychosocial impact of the availability of presymptomatic testing than Huntington's disease (HD), a neurodegenerative disorder. Much of what was initially known about attitudes to genetic testing and the effects that it can have on individuals and their families came from research into HD (Harper, 1997d, p.32-33). Huntington's was the first autosomal dominant, late-onset, severe disorder for which presymptomatic diagnosis was available. The cruel fact that the disease does not usually strike until after the victim has already had children, their early exposure to the full horror of the disease in their own parent, the decades of anticipation of 'the first signs', the insidious onset of symptoms, the distressing cognitive changes and uncontrollable chorea⁷, and the long course of the disease which is inevitably fatal, all combine to make this disease an enormous psychological burden for afflicted families.

A novel by Alice Wexler (1995) shares with the reader an exquisite insight into the dilemmas facing a person living with the threat of HD. While the scientific world now explains this threat thus – that the children of an HD sufferer have a fifty percent chance of carrying the gene and that all of those who do will go on to develop the disease – she indicates that **the emotional and psychological reality of living with this risk feels somewhat different to the risk as it is calculated**. Wexler (1995, p.223) points to a statement made by her sister, Nancy Wexler, on this:

The ambiguous condition of 50% risk is extremely difficult to maintain in one's mind, if not impossible. In practice, a 50-50 risk translates to a 100% certainty that one will or will not develop the disease. (N. S. Wexler, 1979, p.218)

⁷ 'Chorea' is a medical term used to describe "involuntary, excessive, and uncoordinated movement" (A. Wexler, 1995, p.46).

Continuing on this theme, Alice Wexler writes about a particular Venezuelan community afflicted with a particularly high rate of HD who did not need a genetic understanding of the disease to know the emotional effects of it. She highlights the magnificent insight they showed into the psychological burden of being at-risk for an autosomal dominant disease – rather than using the term ‘carrier’ in the way that geneticists eventually would, this community applied it to all those who had a parent with the condition but recognised that only half of these ‘carriers’ went on to manifest physical symptoms. In fact, this way of conceptualising HD acknowledges the ongoing threat to well-being that this disease poses for all those who have been at-risk, whether they turn out to carry the gene itself or not. When presymptomatic testing for the HD gene commenced in other parts of the world, genetic counsellors found that those who tested negative for the gene did not necessarily experience the relief and elation it was anticipated they would (Seymour Kessler, 1994; Tibben et al., 1992; N. S. Wexler, 1992b), as Nancy Wexler has discussed:

It is almost as inconceivable for people to learn that they are not in harm’s way. Identities have been built around being “at risk”: commitments abandoned, lives led in the fast lane. Some people who learn that they are free of the long-dreaded gene are stunned and unprepared. Suddenly they are ordinary; vulnerable now to other diseases, responsible for their lives as never before. Friends and relatives who had sacrificed for them in the past may feel cheated and vengeful or disturbed to find themselves deprived of their role of tending to an invalid. Some people describe “survivor guilt,” especially if they have a sibling or close relative who tested positive for the gene. (1992b, p.2823)

Many of the non-carriers “had shaped their lives against a background burden of HD” and were left feeling that their struggle to “[adapt] to the threat of HD had, after all, been totally unnecessary” (Tibben et al., 1992).

This brings us to another point to consider about genetic testing: its potential to change one’s **sense of identity** – not only in the eyes of the individual tested but also in the eyes of others. It is not just that a person may feel their true identity has been newly revealed to them when a disease gene is uncovered (Armstrong,

Michie, & Marteau, 1998) but also that a person with a 'favourable' test result may feel they have lost part of their identity with their at-risk state no longer being a part of who they are (N. S. Wexler, 1992b). While this challenge to identity exists with any kind of disease, a genetic test reveals information that may influence how we view our ancestry, how we reflect on our own past, how we experience our present and how we look to our future. It has been said that "[r]e-cataloguing illness and pathologies along a genetic axis [...] creates an obligation [for the individual] to act in the present in relation to the potential futures that now come into view" (Novas & Rose, 2000, p.486).

Our genetic make-up is inescapable – we did not catch it by chance, like a virus which may be fought off; we cannot imagine that we are an impersonal host to it and that it is external to us, like a bacterial infection which could have settled with anyone; even a terminal illness, like cancer or AIDS, we were once free from. But our DNA, our genetic make-up, is a timeless and relentless part of us from which there is no escape (for now). It is a unique bequest made to us by our parents. It is an intrinsic part of who we are – in fact, **it was us even before we became who we are**. And, it would seem, what we know about it has the potential to impact on who we think we are...

THE PROBLEM OF KNOWING: POPULATION SCREENING FOR GENETIC CARRIER STATUS

Even when carrier status poses no threat to the individual's own health, discovery of a 'defect' in one's genetic constitution can have profound psychosocial consequences, namely stigmatisation⁸, discrimination and an adverse effect on self-image (Haan, 1993). As stated by Wilfond and Fost (1990, p.2781), "[s]tigmatization can affect a person's self-esteem or have significant social implications if it results in discrimination". This has already been amply illustrated by undesirable outcomes arising from the implementation of mass screening programs in the past, usually

⁸ The term 'stigma' was used by Erving Goffman to refer to an attribute "that is deeply discrediting" (1968, p.13).

where enthusiasm to use new technology has far preceded critical assessment of its safety and effectiveness when used in this context. In particular, the effects of stigmatisation arising from early sickle cell trait (carrier) screening programs, often compounded by confusion, error and inadequate counselling, have been documented. One study in Orchomenos, Greece, revealed that after seven years of sickle cell trait screening, carriers were being socially ostracised because they were seen as undesirable marriage partners, leading many to conceal their carrier status (Stamatoyannopoulos, 1973, p.273). Sickle cell trait screening in the United States in the early 1970s caused many problems for Black communities there. Mandatory sickle cell testing laws were passed in seventeen states, making it compulsory for Black people to be tested for the trait – in some cases, prior to school entrance, and in others, as part of a pre-marital examination (Wilfond & Fost, 1990).

Whitten (1973) voiced early concerns about many aspects of the sickle cell trait screening procedure in the USA, particularly the failure of governments to appropriate necessary funds to implement the mandate (given that many impoverished families were now faced with extra financial burden) and to ensure adequate provision for public and patient education. He warned that “there is no real opportunity for those affected to understand the implications of having sickle-cell trait, or to be relieved of the anxiety and apprehension that this knowledge frequently engenders” (1973, p.318). Testing pre-school children for carrier status was irresponsible and inappropriate given that it would largely only become an issue for them during their reproductive years. It implied that parents needed to be aware of it in the day-to-day care of their child, therefore adding to the misconception that sickle cell trait was a mild form of sickle cell anaemia (Whitten, 1973). This confusion led to stigmatisation of carriers and unfounded discrimination in access to employment, acceptance to the US Air Force Academy, and the ability to obtain health and life insurance (Wilfond & Fost, 1990)⁹. These lessons from

⁹ However, one tends to feel sceptical as to whether this discrimination was based on misunderstanding alone – rather, it may also have provided a convenient excuse for a thinly veiled form of racial discrimination by those with other agendas.

mass screening highlight the psychosocial risks engendered by genetic testing, especially when it is not accompanied by concomitant education and counselling.

Concerns are still being voiced about the possible development of a "genetic underclass of healthy individuals" (Clarke, 1997a, p.150) who are likely to be discriminated against by employers and denied life insurance (and health insurance in some countries) because of a disease susceptibility predicted by genetic testing. If the confidentiality of these test results could be compromised, it is inevitable that employers would be interested in the future health of their employees. The current position adopted by insurers in the UK, USA and Australia is largely that applicants must make available the results of any previously undertaken genetic tests upon request (Harper, 1997b; "Life Insurance and Genetic Testing in Australia," 2002) on the basis of the claim that there is "no significant distinction between genetic information and other health-related information concerning risks" (Novas & Rose, 2000, p.499). Although insurance applicants are not required to undergo testing at the moment, one of the greatest concerns for genetics professionals is that "the use of results to alter risk calculations could become a pressure for individuals to be tested who might not otherwise have wished for this" (Harper, 1997b, p.54). Conversely, others may be deterred from a test they wish to undergo for personal and reproductive reasons for fear of adverse insurance consequences if they receive a high-risk result. There is also the danger that ignorance regarding the health status of carriers of recessive disease will result in unjust discrimination. As an example, Harper (1997b, p.53) points to the results of an American survey of insurance companies in the early 1990s¹⁰ where over half considered the carrier state for conditions such as cystic fibrosis and Tay-Sachs disease to be a 'pre-existing condition'.

So, most importantly, it must be emphasised that the risks of stigmatisation and discrimination associated with genetic screening are still very real threats today and

¹⁰ US Congress Office of Technology Assessment (1992).

should not be viewed as relics of a less enlightened past. Wilfond and Fost (1990, p.2778) recommend that:

Mass genetic screening programs should be considered experimental public health programs, implemented only after a favorable assessment of a program design that effectively achieves its goal while minimizing the potential medical, ethical, legal, and social problems.

Harper and Clarke (1997, p.75) too warn of the “potential for harm caused by a programme of genetic services whose focus is on the population rather than the individual”, reminding us that there is a great difference between family-based genetic counselling and population-based genetic screening – the former entails responding to pre-existing concerns in a family already struck by a particular disorder, while the latter involves raising anxieties among large groups in the population who may have little to no knowledge of the disorder in question (Clarke, 1998, p.4).

Clarke (1997c, p.84-85) asks us to consider the collective-social consequences of population carrier screening programs. He suggests that society may become less tolerant of genetic disease if it is seen as potentially ‘preventable’, undermining respect for affected individuals and reinforcing the stigmatisation of these people and their families. Individuals may feel pressured to undergo carrier testing and may be seen as irresponsible and blameworthy if they give birth to a child with a costly genetic disorder when there were options available to avoid this. The burden of imposing genetic decision-making on whole communities will be felt most heavily by mothers – those who could face being socially ostracised if they decide not to terminate a much-wanted pregnancy, or lifelong guilt and sadness if they do. We must understand that **eliminating** genetic disease by carrier testing, prenatal diagnosis¹¹ and termination of affected pregnancies is not equivalent to **preventing** genetic disease and the profound and devastating impact it can have on families¹².

¹¹ Lippman defines ‘prenatal diagnosis’ as referring to “all the technologies currently in use or under development to determine the physi(ologi)cal condition of a fetus before birth” (1991, p.19-20). These techniques include ultrasound screening, chorionic villus sampling and amniocentesis. According to Lippman, prenatal diagnosis techniques play a part in the process of geneticisation

WHAT IS GENETIC COUNSELLING?

The current philosophy of evidence-based health care and practice demands that the counselling aspects of the genetic testing process receive as much research attention as the laboratory aspects. Knowledge of the objectives of a genetic counselling service is essential for its evaluation but published definitions and guidelines about the objectives of genetic counselling vary and have been shifting over time (Michie, Smith, Heaversedge, & Read, 1999). Some have emphasised the provision of information only: "Genetic counselling is the communication of information and advice about inherited conditions" (Connor & Ferguson-Smith, 1993, p.113). According to Clarke (1998), this definition reflects the practice of many clinical geneticists. Others have seen it as a communication and psychotherapeutic process (Biesecker & Marteau, 1999; N. S. Wexler, 1979), in which counsellors "help clients reach decisions, deal with couple's conflicts (*sic*), manage guilt and shame, provide empathy, and, in general, maintain a nondirective stance" (S. Kessler, 1997, p.294). In a report based on a workshop organised under the sponsorship of the National Genetics Foundation, Inc. in Washington, D.C., in 1972, genetic counselling was defined as "a communication process which deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family" (cited in Fraser, 1974, p.637), adding more specifically that the counsellee(s) should be helped to make decisions which seem most appropriate to them, and to make the best possible adjustment to their genetic situation. This definition is compatible with the findings of a study conducted over twenty years later that identified the five most frequently cited and highly ranked objectives of genetic service providers as, in descending order, to (1) provide information, (2) give support, (3) facilitate decision-making, (4) assess risk, and (5) achieve understanding (Michie et al., 1998).

(1991, p.21). She says while "language of control, choice and reassurance" has been "successful as a marketing strategy", prenatal diagnosis is undeniably "a means of separating fetuses we wish to develop from those we wish to discontinue. Prenatal diagnosis does approach children as consumer objects subject to quality control" (1991, p.23).

¹² This is discussed later in Chapter Seven (see section: Technology Does Not Eliminate Suffering).

The most recent guiding statement to genetic counsellors in Australia defines their practice as “a communication process, which aims to help individuals, couples and families **understand and adapt** to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions” (“Australasian Society of Genetic Counsellors (ASGC) Code of Ethics,” 2008, p.1). Of note here is the silence around what ‘adapting’ to these implications entails. Novas and Rose are more explicit in their ‘definition’, saying

the practices of genetic counselling **incite** an individual, couple or family to reflect upon their genetic constitution with the aim of affecting their conduct in light of this knowledge. [emphasis added] (2000, p.492)

In other words, there is an expectation that people will **act** in relation to the genetic information they now have about themselves; that they will **assume responsibility** for the management of the genetic threat now known to them (Novas & Rose, 2000).

A study which investigated clinical geneticists’ thoughts and beliefs about their practice identified a great range of complex psychological tasks seen as necessary to genetic counselling, each with a contradictory aspect (Michie et al., 1999). These were listed as working out an appropriate stance in terms of:

1. Providing information that is objective, full, and accurate versus information that is contingent on circumstances and tailored to individual needs;
2. Eliciting emotion and dealing with it directly versus dampening down and containing emotion; and
3. Communicating non-directively versus directly.

The authors suggest that these main tensions in the tasks of genetic counselling arise because the genetics professionals are influenced by two competing models of practice, previously examined and contrasted in a paper by Seymour Kessler (1997). Kessler delineated the goals and underlying assumptions of each model in two separate tables (1997, p.288/290), which have been replicated exactly on the next page:

Table I. The Teaching Model

-
1. Goal: educated counselees
 2. Based on perception that clients come for information
 3. The model assumes that if informed, client should be able to make their own decision
 4. Assumptions about human behavior and psychology simplified and minimized; cognitive and rational processes are emphasized
 5. Counseling task is to provide information as impartially and as balanced as possible; correct misinformation
 6. Education is an end in itself
 7. Relationship with client based on authority rather than mutuality
-

Table II. The Counseling Model

-
1. Goals
 - a. to understand the other person
 - b. to bolster their inner sense of competence
 - c. to promote a greater sense of control over their lives
 - d. relieve psychological distress, if possible
 - e. to support and possibly raise their self-esteem
 - f. to help them find solutions to specific problems
 2. Based on perception that clients come for counseling for complex reasons (e.g., information, validation, support, anxiety reduction)
 3. The model has complex assumptions about human behavior and psychology which are brought to bear in counseling
 4. Counseling tasks complex
 - a. requires assessment of client's strengths and limitations, needs, values and decision trends
 - b. requires range of counseling skills to achieve goals
 - c. requires individualized counseling style to fit client's needs and agendas; flexibility
 - d. requires counselor to attend to and take care of his/her own inner life
 5. Education is used as a means to achieve above goals
 6. Relationship aims for mutuality
-

Kessler had explained that the teaching model "attempts to end up with an 'educated' counselee" while the counselling model "hopes to leave the [counselee] psychologically more autonomous and functional" (1997, p.287). The teaching model asserts the necessity of providing full objective information and correcting misinformation and misperceptions but unfortunately this is seen as incompatible with attending to emotional concerns, seemingly because emotionality is perceived as a hindrance to the learning process. The counselling model recognises that

people's motivations for seeking genetic counselling are complex and varied, usually exceeding the requirement of information alone. This model demands engaging with and understanding the counsellee's "strengths and limitations, needs, values, and decisional trends" so that self-efficacy, self-esteem, psychological, emotional and decision-making issues can be addressed if need be (S. Kessler, 1997, p.290). Their perceived needs must be assessed so that information can be tailored to their personal circumstances¹³.

While trying to accommodate two competing models of practice would certainly bring dilemmas in terms of juggling the contradictory aspects of counselling, I suggest a major contributing factor to this difficult aspect of genetic counselling is the dilemmas inherent in the very nature of human existence itself which all come into play in the genetic testing arena. This is in line with what professor of law, Dena Davis, has said:

[W]e can see that genetics encompasses just about all of the most emotionally powerful of human experiences: life, death, reproduction, parenthood, and the meaning of health and disease. (Davis, 2001, p.12)

I believe this is what makes the impact of genetic testing on families so amenable to an investigation driven by the tenets of existential philosophy.

ETHICAL ISSUES FACING GENETIC COUNSELLORS: DEALING WITH PAST, PRESENT AND FUTURE

Considered to be one of the central tenets of genetic counselling, adherence to the principle of **non-directiveness**¹⁴ derives from an ethos that emphasises the

¹³ It has previously been suggested that giving personally tailored information in genetic counselling sessions may be a more successful strategy than trying to provide all potentially relevant information on the basis that medical and technical information provided may be recalled less well than the implications of that information for the family (Michie, Marteau et al. 1997; Michie, McDonald et al. 1997; Michie, Smith et al. 1999).

¹⁴ The concept of non-directiveness does not appear in the ASGC Code of Ethics (2008), however, 'non-coercion' is mentioned, albeit in the Glossary only, in relation to obtaining informed consent. This point will be returned to later in this chapter (see section: Is Non-Directive Genetic Counselling Possible?).

autonomy¹⁵ of the counsellee and their right to make decisions which have not been imposed by the counsellor (Clarke, 1998, p.5). As previously articulated by Clarke, "[t]he concepts of autonomy, informed consent and non-directiveness are closely interwoven" (1998, p.5). Respect for the autonomy of an individual requires that **informed consent**¹⁶ is obtained before any genetic test is performed. The provision of complete information regarding various options available to the counsellee is therefore an integral part of sound genetic counselling and it is necessarily crucial that these options are presented in a non-directive manner if counsellees are to make their own, autonomous decisions.

Another aspect of respecting autonomy is respecting that the individual has ultimate control over their own personal genetic information. This presents great ethical and practical challenges to genetic counsellors when a client wishes to withhold genetic information from other family members who may find it extremely valuable (Clarke, 1997a, p.152-157). While one is sympathetic toward genetic counsellors who feel that in these circumstances they have a duty to break their client's **confidentiality**, it has previously been suggested that this action could ultimately undermine the community's confidence in their credibility and integrity as health professionals and weaken medical respect for confidentiality in general (Clarke, 1997a, p.152-153). Despite this, it would seem that attitudes towards the disclosure of genetic information to genetic relatives could now be shifting. In an inquiry into this issue by the Australian Government ("Australian Law Reform Commission Report No. 96: 'Essentially Yours: The Protection of Human Genetic Information in Australia'," 2003), this issue was referred to as "a looming area of medico-legal controversy"¹⁷ (section 21.19). Despite acknowledging that "[t]he overriding responsibility of the clinical geneticist remains with the patient and not

¹⁵ In the ASGC Code of Ethics, 'autonomy' is defined as "[t]he ability to be self-determining and self-governing; the capacity to make reasoned choices on the basis of adequate information" (2008, p.4).

¹⁶ The ASGC define 'informed consent' as "[c]onsent obtained on the basis of the non-coercive provision of adequate current, relevant, balanced information, which can be readily understood by the person expected to give consent. The person to give consent must also have been afforded adequate time and opportunity to consider the information, seek clarification from other sources or support persons before making their decision" (2008, p.5).

¹⁷ On this point, the report cites Bell and Bennett (2001).

to any other family members”¹⁸ (section 21.19), the Inquiry concluded that current privacy legislation in Australia “inappropriately constrains health professionals’ decisions about the disclosure of clinically relevant information to genetic relatives” (section 21.22). In order to “remedy this situation”, the Inquiry proposed amendments to the legislation and the development of National Health and Medical Research Council (NHMRC) guidelines on this issue (section 21.22)¹⁹.

Another ethical dilemma which challenges genetic counsellors is the situation that confronts them when the **wishes/demands of parents** regarding testing of their child seemingly conflict with the **rights of that (future) child**. A child cannot be said to give informed consent for a procedure that they may not fully understand. The genetics professional must “guard the privacy of those whose personal information is mined without their knowledge or consent”, as is the case with the child or foetus whose genotype is tested (Davis, 2001, p.79). Davis makes comment on the rights of the child to **privacy**:

[I]t appears that the right of privacy the child will have when she becomes an adult, to decide what medical and genetic information she will choose to share with her family, should be respected now and not sabotaged by allowing her parents to have her tested (Davis, 2001, p.76).

This is especially so when the genetic information is not necessary for the child’s health and welfare, as is the case with carrier status for recessive diseases where

the most important use of the information is to make marital and reproductive decisions in adulthood. These decisions—whether to marry, whether to have children, whether to make use of controversial procedures to avoid genetic risk—are clearly within the child’s most protected zone of privacy once she becomes an adult (Davis, 2001, p.80).

¹⁸ Here, the report cites Pergament (1997).

¹⁹ In response to this, privacy legislation was amended in 2006 and, in 2008, the NHMRC released draft guidelines for public consultation, which address the circumstances in which disclosure by health practitioners of genetic information to genetic relatives becomes legally justifiable. Reference: NHMRC n.d. *Public consultation on the ‘Disclosure of genetic information to a patient’s genetic relatives under section 95AA of the Privacy Act 1988 (Cth) – Guidelines for health practitioners in the private sector’*. Retrieved 10 March 2009, from: http://www.nhmrc.gov.au/guidelines/consult/consultations/disclosure_genetic_info.htm

However, a potentially even greater and more sinister threat to the genetic privacy of the individual – not just children – inevitably comes from powerful third party entities. Genetics professionals need to contend with other major stakeholders in the genetic constitution of the individual, such as the state, biotechnology corporations, employers, and insurance companies.

The potential clearly exists for the systematic abuse of information stored in genetics records and registers. **Eugenic²⁰ abuses** of human rights are the most heinous example. Most disturbing in the context of the current discussion is that past compulsory sterilisation laws in the United States and Nazi Germany were direct applications of eugenic views espoused by some of the very scientists and medical professionals researching and ‘treating’ the conditions included in the legislation (Harper, 1997c, p.231-232). By his own report²¹, and well aware of the fate in store for those in his ‘care’, at least one doctor actually passed on to Nazi authorities those patients he had diagnosed with HD and their at-risk siblings and offspring (cited in Harper, 1997c, p.232). Had computerised genetic registers and more sophisticated genetic tests (e.g., presymptomatic test for HD) existed at that time, they would undoubtedly have been exploited in the name of eugenics by the Third Reich (Harper, 1997c, p.232-233). It would be remiss to assume that this kind of barbarism could never recur in present times.

The “first clearly eugenic law” in the world of modern genetics – that of “Maternal and Infant Health Care” – has already been introduced in China (Harper, 1997a, p.237). Some of the clauses in the law that are specifically relevant here are cited (from the official Chinese translation into English) in Harper’s work (1997a, p.237). Since 1995, the Chinese Government has required that “genetic diseases of a serious nature”, among other diseases, be included in a pre-marital physical check-

²⁰ While the word ‘eugenics’ was originally used to denote the discipline of applied human genetics, the term has become an emotive one with “overwhelmingly negative connotations” as a result of the abuses of human genetics that occurred in the first half of the twentieth century (Clarke, 1997d, p.182). The term is now synonymous with the practice of (forcibly) subordinating or sacrificing “the rights and welfare of individuals [...] to the supposed interests of collective entities such as society, the state, the race or the species” (Clarke, 1998, p.1).

²¹ Harper cites Panse (1942).

up (Article 8, s1). If the man or woman is diagnosed with such a genetic disease, the two cannot be married unless “both sides agree to take long-term contraceptive measures or to take ligation operation for sterility” (Article 10). If a married couple of childbearing age is afflicted with a genetic disease, they shall comply with “medical advice” (Article 16), and if prenatal diagnosis reveals a foetus has a genetic disease or a defect of a serious nature, the parents shall be directed to terminate the pregnancy (Article 18, s1 & s2). An official commentary on the draft version of this law stated that it will allow ‘China’ to “use legal means to avoid new births of inferior quality and heighten the standards of the whole population” (cited in Harper, 1997a, p.239). Even more alarming than this, however, was the casual approach used by the commentator in pointing out that “[t]he draft does not state whether China will adopt euthanasia to eliminate congenitally abnormal children [because] the international community has not come to a conclusion on that issue” (cited in Harper, 1997a, p.239).

IS NON-DIRECTIVE GENETIC COUNSELLING POSSIBLE?²²

It is easy to feel shocked and disgusted by the existence of the “Maternal and Infant Health Care” legislation in China today ... until those feelings are surpassed by a realisation that the political, economic, professional and social contexts in which most other genetic services around the world are set must ultimately produce net results which are not dissimilar to the results of the Chinese law²³. Troy Duster aired early concerns about this in his book, *Backdoor to Eugenics* (1990), which Clarke summarised well:

There also exist less explicit forms of eugenics in which the forces of the ‘free’ market, aligned with social convention, may achieve the goals of the eugenics movement more effectively than it managed to do for itself (Clarke, 1998, p.1).

²² A question posed by Clarke (1991).

²³ In fact, one study that looked at genetic counselling from multiple international perspectives found evidence that genetic service providers in certain South American countries “openly strive to reduce the incidence of affected births”; and that within Europe the “slant of ‘directiveness’ varies”, one example being that termination of affected pregnancies are more likely to be encouraged by Portuguese than by German geneticists (Biesecker & Marteau, 1999, p.133).

Duster predicted that the power of technological advances would bring with it a new attitude – that “the defective fetus” can be eliminated (1990, p.128). It would be difficult to argue that this has not turned out to be the case in the 2000s, given that – in his words (1990, p.128) – “the elimination or prevention of the ‘defective fetus’ is the most likely consequence and ultimate meaning of a genetic screen” today. Concurring with sentiments expressed in another commentary²⁴, Duster (1990, p.145-146) said that offering genetic counselling at all provides implicit endorsement and “a new kind of legitimacy” to the attitude that elimination of the defective foetus is desirable.

The ethos of non-directiveness has been widely promulgated within genetic counselling circles by those who understandably wish to distance themselves and their practice from past and present eugenic abuses of human rights but Angus Clarke appears fully justified in questioning whether non-directive counselling can be achieved in the genetic testing context at all. He has suggested that the very fact that:

prenatal diagnostic and screening tests are available imposes a burden of responsibility on every couple embarking upon a pregnancy, whether at increased or standard risk of having a child with a serious problem [and this] will lead some individuals to feel pushed, against their wishes, to accept whatever offers of prenatal testing are made (Clarke, 1997d, p.181).

This is cause for reflection for genetics professionals around the world who regard the environment in which they currently offer counselling as non-directive and one in which counselees are able to make fully autonomous decisions.

Perhaps the answer is to move away from the use of ‘non-directiveness’ as the catchcry for genetic counsellors. Seymour Kessler suggested alternative ‘ideals’ in his discussion of the two models for genetic counselling:

Under the teaching model the professional tries to achieve **neutrality, even-handedness, impartiality, and noncoerciveness,**

²⁴ Duster refers to the work of Hsia, Y. Edward, *et al.* (1979).

and these terms may be more appropriate as ideals and more descriptive of the procedures in the model than the more psychologically-loaded term, nondirectiveness [emphasis added] (1997, p.289).

One would think it equally desirable that ideals such as these be embraced by the professional using the counselling model also. As with non-directiveness, the principles of value-neutrality²⁵, even-handedness, impartiality and non-coerciveness also emphasise the autonomy of the counsellee and their right to make decisions which are their own and have not been imposed by the counsellor.

Still, it remains difficult to envisage a couple achieving true autonomy when making the 'informed reproductive decisions' which are said to be facilitated by genetic testing and counselling given that reproductive plans and behaviour are so often used as measures of the 'effectiveness' of genetic counselling services. In this context, 'effective genetic counselling' can only be construed as that which **leads** clients to make the **right** decision – to terminate any pregnancy if the child is "likely to cost more in medical care and social benefits than he/she would pay in taxes over his/her lifetime" (Clarke, 1997b, p.169). The fact that clinicians are pressured to justify the provision of genetic services in terms of financial savings, when "the provision of all other aspects of clinical medicine is accepted as costing money" (Clarke, 1997b, p.169), is a strong indication that there are powerful economic and sociopolitical structures providing us with an environment in which we are **allowed** to make our informed reproductive decisions. It is important that people have the freedom to make decisions which are best for themselves and not for the collective conscience or the collective purse.

²⁵ The notion that value-neutrality should be included in a model for genetic counselling has been challenged. Davis says the ethos of value-neutrality affirms that "a decision's moral worth is determined solely by whether it is the right decision for [the] client" (2001, p.15). She refers us to a paper by Wachbroit and Wasserman (1995), in which they surmise that value-neutrality seems to be valued primarily because it is thought to be a necessary condition for client autonomy. What underpins this assumption is "a belief that the expression of the counselor's values undermines patients' abilities to make decisions for themselves" but Wachbroit and Wasserman say this assumption is unwarranted and argue that, in fact, value-neutrality is "neither desirable nor possible" (1995, p.104). Davis summarises their challenge: "[They argue] that clients are not that fragile, and also that unspoken social biases may be more threatening to autonomy than those that are clearly put on the table" (2001, p.21).

The testing technologies that allow for carrier testing, prenatal diagnosis of an affected foetus (or even pre-implantation diagnosis of an affected embryo), and consequent continuation or termination of a pregnancy, raise huge ethical, moral and, potentially, legal^{26,27} dilemmas for couples, clinicians, and society.

Angus Clarke (1998, p.1) has referred to those identified as presymptomatic carriers of an untreatable disease as having “knowledge without power”. However, the ‘knowledge with power’ which presumably arises when a healthy carrier of a recessive disease has access to prenatal diagnosis is not necessarily empowering. The scepticism about whether purely non-directive genetic counselling can be achieved, the perceived expectations of society, and the spectre of eugenics, raise many questions and concerns about the quandary in which these newly-available ‘choices’ place prospective parents.

So how do parents feel when they discover this ‘quandary’ awaits the adult who their child is-to-become?

GENETIC TESTING OF CHILDREN

What seems to emerge from much of the rhetoric surrounding genetic testing today is a belief that, rather than carrier testing (which is the focus of this study), it is “in the category of ‘presymptomatic’ and ‘predictive’ testing that most of the difficult

²⁶ Margery Shaw, a prominent geneticist and lawyer working in the United States, has written on the topic of “whether or not a defective fetus should be allowed to be born” (1984, p.1). She talks about the various situations in which there is ongoing potential for ‘wrongful birth’ and ‘wrongful life’ claims to be brought before the courts and quoted one judge from the California intermediate appellate court as stating: “If a case arose where ... parents made a conscious choice to proceed with a pregnancy, with full knowledge that a seriously impaired infant would be born ... we see no sound public policy which should protect those parents from being answerable for the pain, suffering, and misery which they have wrought upon their offspring” (1984, p.8). In this situation, a tort action known as wrongful life may allow a child to bring a law suit against their parents, claiming that they should never have been born at all (N. S. Wexler, 1989).

²⁷ In Australia, a landmark legal action was recently launched against a Victorian IVF clinic, alleging staff were negligent in implanting an embryo with a hereditary cancer gene that had supposedly been screened out using pre-implantation diagnosis. Reportedly, the parents did not find out their son carried the gene until three months after his birth and are now suing for “damages to cover hospital and medical expenses for their son’s entire life, [...] special damages for the cost of bearing and rearing another child without the mutant gene [...] [and] compensation for emotional pain and upset” (Hudson, 2008).

issues involving genetic testing lie” (Harper, 1997e, p.9). In one particular discussion (Harper, 1997e, p.11-12), the importance of the pre- and post-test counselling aspects of presymptomatic testing for late-onset disorders was emphasised, while these aspects of cystic fibrosis carrier testing were referred to as “relatively simple” and only needing to be “quite limited” (so much so that written information was thought to be an appropriate replacement for counselling).

This stance belies the ‘difficulty’ of the issues involved with testing for healthy carriers. It completely underestimates the emotional burden imposed on parents knowing that they could be ‘responsible’ for passing on a hereditary disease to their child. It overlooks the complex decisions that the individual identified as a ‘carrier’ may be confronted with in light of the new reproductive ‘choices’. In particular, it forgets the **complexities** and **sensitivities** of pre- and post-test counselling where the **carrier testing of healthy children** is concerned. This practice has been described by various commentators thus: as “ethically worrisome”, particularly where “the information is not immediately useful for the child’s reproductive decision making” or “if the request is from the parents rather than the child” (Wertz & Reilly, 1997, p.1163); as “unnecessary and unethical” (Harper & Clarke, 1990, p.1206); as warranting a “cautious approach” (Clarke, 1998, p.7); as “not appropriate” (Davis, 2001, p.85); and also as “the most ethically problematic category of [genetic] testing” (Wertz, Fanos, & Reilly, 1994, p.879).

Knowledge of the consequences of performing carrier tests on children is scarce. **Suppositions** about the potential harms of this kind of testing during childhood abound, and variously include: the possibility of stigmatisation, reinforcement of latent feelings of unworthiness, that misunderstandings by parents or child could lead to serious misconceptions about their future, ‘survivor guilt’²⁸, blame, erroneously-altered perceptions of risk in untested siblings, fears related to intimacy and interpersonal relationships, harm to the parent-child bond and harm to the child’s self-concept (summarised by Wertz et al., 1994, p.876). In contrast,

²⁸ We will re-visit this concept, and come to better understand it, in Chapter Six (see section: The Children Who Are ‘Only’ Carriers).

the potential benefits seem to be limited to reduction in parental anxiety (when they are the ones requesting the test) (Wertz et al., 1994, p.878) or otherwise the opportunity to gently introduce the genetic information to the child over the course of their development (Fryer, 2000, p.284).

Due to concerns that carrier testing may be contrary to the best interests of the child, most concur that where there is no current reproductive benefit it is not generally advisable to perform carrier tests for recessive disorders on children²⁹. Although there is no clear consensus as to when testing should be deferred to, some recommendations have been: until the individual is of an age where they are “able to consider their own future reproductive plans” (Harper & Clarke, 1990, p.1206); “until adulthood” (Wertz et al., 1994, p.879); “until the child is able to understand the issues and request testing in person” (Fryer, 2000, p.285); and “until the child has the intellectual capacity needed to discern if and when to be tested” (Borry, Fryns, Schotsmans, & Dierickx, 2006). While these alternate viewpoints are each inherently nebulous, and potentially allude to quite a difference in age/maturity, we suspect the authors share a common **concern for autonomy over decision-making**. It is ethically problematic to deny the future autonomy of the child in favour of the present autonomy of the parents – once carried out, testing forecloses the possibility that the adult who the child is-to-become may choose not to seek out this genetic information. This topic will be explored further in Chapter Four (see section: Preserving the child’s right to an open future).

Despite these concerns, there are situations in which parents are coming to know that their children carry a recessive disease gene. Some parents become aware of their child’s carrier status through a request for prenatal or childhood testing³⁰ due

²⁹ Unfortunately, the recently-revised ASGC Code of Ethics (2008) makes no reference to the issue of genetic testing of children at all. The Human Genetics Society of Australasia does have a position statement on presymptomatic and predictive testing in children and young people but, as at March 2009, have not issued a corresponding document in relation to carrier testing in children and young people (see ‘Policies, Guidelines, Statements’ at <http://www.hgsa.com.au/>).

³⁰ This is contrary to recommendations that carrier tests not be performed on children. One study in the USA confirmed that a majority of DNA diagnostic laboratories are responding to parental requests for carrier-testing of children under twelve years of age (Wertz & Reilly, 1997). Another in the UK found the main reason given by health /genetics professionals for performing carrier tests on

to a family history of the disease. In one paper in a paediatric nursing journal, parents of a child with CF were specifically mentioned as those who may want their other children tested to determine if they could be carriers (Williams, 2000, p.209). Also with reference to the cystic fibrosis gene, there are even parents who may never have before heard of the disease who are discovering their child is a carrier. This is not through actively seeking out this information but as a result of neonatal screening programs which have been implemented at children's hospitals for the early detection of babies with CF. By virtue of the way the two-tier screen is conducted, the incidental detection of a small proportion of healthy carriers is unavoidable (Ranieri et al., 1994). Very little is known about what this genetic information means to parents who become the custodians of the information and wholly responsible for divulging it to their children as and when they see fit³¹.

Due to the ethical dilemmas which face all those who must make decisions in the best interests of the child, the genetic testing of children remains an extremely controversial topic. While debate has been thought-provoking and provided insight into the possible ramifications of testing, much is based on the beliefs and opinions of those who must necessarily be influenced by their own personal and professional interests, experiences and prejudices³². While many continue to speculate about whether the genetic testing of children is ethical, where it only has application to

children was "parental pressure to test" (Fryer, 2000, p.283). We will also see that at least one participant in this study (Rose) had her request acceded to for her child to be carrier-tested (although it should be noted that she was living in the UK at the time).

³¹ One Australian study (based solely on postal questionnaires) found that, ten years post-neonate screen, almost all parents remembered that their child had been identified as a carrier but some (28%) had residual anxiety about the current health of their carrier child and about potential difficulties their child might have in future relationships and with reproductive decision-making (Lewis, Curnow, Ross, & Massie, 2006). Another small study in the UK used a mixed-methods approach to investigate the psychosocial implications for these families (Parsons, Clarke, & Bradley, 2003). They found that all were in favour of the neonatal screening program and "there was no evidence that the mother/baby relationship, maternal anxiety, or well-being had been adversely affected" (2003, p.F470).

³² For instance, professional-society statements point to threats to the child's autonomy while consumer groups can tend to favour the parents' rights to testing (Wertz & Reilly, 1997). The latter argue that "parents are better able to predict the psychosocial outcomes of testing than are physicians" and that carrier testing "may be less traumatic in childhood than in adolescence (when the minor may become reproductively active and may seek testing anyway) and that such testing may reduce anxiety and is not stigmatizing if the family is educated sufficiently, through counseling" (Wertz & Reilly, 1997, p.1165).

their reproductive futures, it has been noted that there is little empirical evidence available to indicate whether testing of this kind is beneficial or harmful³³ (A. Clarke, 1998; Fryer, 2000; Wertz & Reilly, 1997; Williams, 2000). This question is set to become even more pertinent if the frequency at which children are being identified as carriers increases. This is sure to occur if calls to replace newborn screening with population screening for CF carriers³⁴ are heeded, as this will increase the uptake of prenatal tests for CF and, hence, the overall numbers of parents who discover their unborn baby is 'only' a healthy carrier.

The alleviation of uncertainty about an individual's carrier status early in life may be beneficial with regard to their ability, and that of their parents, to adapt to this information. Alternatively, there may be a real potential for harm to the child's psychological health (due to stigmatisation by themselves or others) if this knowledge is imparted before they are emotionally prepared to accept it or in such a way that they feel their rights to privacy and autonomy have been diminished. Notwithstanding the advice not-to-test, as has been discussed, the reality is that there are a variety of situations in which parents are discovering that their children carry a recessive disease gene. They then become the caretakers of this genetic information, entrusted to pass it on to their children as and when they see fit.

How can we better understand the outcomes of genetic testing for carrier status as a foetus; as a neonate; as a child? Research is required with the invaluable group of families where a child's carrier status is already known to their parents. The case of the cystic fibrosis gene is particularly suited to an inquiry of this kind because there are a variety of situations in which parents are coming to know that their healthy children are carriers of the gene. What can be discovered by

³³ In one exploratory study, Jolly, Parsons, and Clarke (1998) assessed ten families where children had been identified as carriers of a balanced chromosomal translocation and who were now over ten years of age. They found that the individuals did experience "a disruption of their psychological equilibrium" (i.e., initial shock, anxiety and some feelings of being stigmatised) when they discovered their carrier status but that this was "transient" (1998, p.87). None reported feeling resentful about being tested as a child and some were, in fact, glad that this had occurred when they were younger.

³⁴ A number of papers in the recent Australian literature have advocated this (J. Massie, Forbes, DuSart, Bankier, & Delatycki, 2007; McClaren, Delatycki, Collins, Metcalfe, & Aitken, 2008; Wake et al., 1996).

learning more about these families and their experiences to date? This thesis is devoted to the answering of this question.

INTRODUCING CYSTIC FIBROSIS (CF) AND ITS 'CARRIERS'

Cystic fibrosis (CF) is a monogenic disorder that presents as a multisystem disease (Boucher, 2008). Individuals with cystic fibrosis have two loss-of-function mutations in a gene located on chromosome 7, which encodes the CF transmembrane conductance regulator (CFTR) protein. This protein forms a chloride channel for epithelial membranes (Woolcock, 2006). All tissues that express the CFTR protein express abnormal ion transport function in CF patients but the effects this has on electrolyte and water transport are organ-specific (Boucher, 2008).

The definition and diagnosis of CF has been evolving since the 1920s, when the familial nature of a 'congenital intestinal obstruction' was first documented by the Swiss pædiatrician, Guido Fanconi (Kerr, 2005). Anne Kerr has presented an excellent sociohistorical analysis of the evolution of this disease, and her summary of cystic fibrosis as we know it today is cited here:

CF is now thought to be one of the most common genetic conditions, affecting approximately 1:2,500 newborns. Typically, it is characterised by chronic obstruction and infection of the lungs³⁵ and defective function of the pancreas, which causes digestive problems³⁶. The clinical diversity of CF, however, means that the time of onset and severity of pancreatic and the pulmonary disorders that it involves vary³⁷. An autosomal recessive mode of inheritance for the condition was first suggested in 1946 and confirmed in 1949. This means CF is inherited from two unaffected 'carrier' parents (the frequency of

³⁵ This ultimately leads to bronchiectasis and bronchiolectasis, which cause localised irreversible dilation of parts of the bronchial tree (Boucher, 2008).

³⁶ Exocrine pancreatic insufficiency affects more than 90% of patients with CF (Boucher, 2008). The inadequate pancreatic secretion of digestive enzymes in these patients leads to protein and fat malabsorption, as well as the malabsorption of fat-soluble vitamins.

³⁷ It has been suggested that this is due to environmental factors and 'modifying genes', other than the CFTR gene (Ratjen & Döring, 2003, p.681).

the carrier rate in the population is typically estimated as 1:25). The life expectancy of people with CF has risen to around 35 over the course of the 20th century, and people with CF are now involved in a range of adult associations with various links to CF charities. Following advances in recombinant DNA and sequencing technologies, and the development of new techniques like chromosome walking and jumping, the gene for CF was identified in 1989. Since then over 1,300 different ways this gene can mutate have been identified³⁸, but the most common one – delta F508³⁹ – is thought to account for over 80 per cent of CF cases in what is described as the Caucasian population. (2005, p.875)

Those people whom Kerr refers to as ‘carriers’ have only one mutation in the CFTR gene⁴⁰ – this does not affect their health in any way as the normal copy of the gene compensates for the loss-of-function in the faulty one. When two carriers of a recessive disease have a child together they have a one-in-four chance in each pregnancy that the baby will be affected, a one-in-two chance the baby will be a carrier like the parents and a one-in-four chance the baby will have two normal copies of the gene, therefore being neither a carrier nor affected.

Diagnosis of CF has traditionally been based on clinical features and confirmed by the finding of an elevated sweat chloride concentration but since 1989 it has been possible to also use gene mutation analysis to aid the diagnosis (Mishra, Greaves, & Massie, 2005). Signs and symptoms of CF usually appear in early childhood but newborn screening for the disease means that in most cases it can now be aggressively and pre-emptively treated from birth⁴¹. It is believed that this early treatment is important in determining subsequent clinical outcomes for children with CF and that positive gains made are still apparent in adolescence (McKay,

³⁸ As at March 2009, this figure had climbed to over 1600 mutations (for up-to-date information, see the Cystic Fibrosis Genetic Analysis Consortium webpage: <http://www.genet.sickkids.on.ca/cftr/>).

³⁹ This is caused by a deletion of phenylalanine at position 508 in the CFTR protein. While it is the most common mutation world-wide, its frequency varies between ethnic groups (Ratjen & Döring, 2003, p.681).

⁴⁰ Hereafter this gene will generally be referred to as ‘the CF gene’ in this thesis.

⁴¹ Treatment for CF consists of a once- or twice-daily routine of chest physiotherapy to clear infected secretions, use of antibiotics to treat infections, a diet with a high fat intake, and oral pancreatic enzyme supplements with every meal (Woolcock, 2006).

Waters, & Gaskin, 2005; Waters et al., 1999). Newborn screening for CF has been occurring in Australia for over twenty years (R. J. Massie & Clements, 2005), with the two-tier neonate screen for the disease being introduced at various hospitals across the country in the early 1990s (shortly after the CF gene was identified) (Massie, Olsen, Glazner, Robertson, & Francis, 2000; Ranieri et al., 1991; Wilcken, Wiley, Sherry, & Bayliss, 1995).

The first tier of the screen involves measuring immunoreactive trypsinogen concentration in dried blood spot samples from neonates aged 3-5 days (Ranieri et al., 1994). The blood is obtained from a heel prick of the newborn and is known as 'the Guthrie test'. In the second tier, direct gene analysis to detect the more common cystic fibrosis mutations (deltaF508, deltaI506, G551D, G542X and R553X) is performed in those blood spot samples which produced the highest 1% of immunoreactive trypsinogen⁴² values, and also in those from all neonates with meconium ileus or a family history of CF (Ranieri et al., 1994). The newborns with two identifiable mutations are referred for immediate clinical assessment and confirmatory sweat test, while those with one identifiable mutation are recalled for a sweat test when they are about three to four weeks old. This is necessary because they may have another less common CF mutation that was not tested for in the screen.

At this stage, the parents are advised by telephone of their child's at-risk status for CF and informed that a sweat test is required to determine whether the child is a healthy carrier or affected with the disease. The parents of these babies are offered genetic counselling and the option of carrier testing also, as it is now known that at least one of the child's parents must be a CF carrier also. Of course, this is a very stressful time for the parents – the news comes 'out of the blue', they worry tremendously as they await the appointment, and then bear witness to their suddenly-vulnerable newborn undergo a medical procedure that is, in this context, now portentous of the life their child may have ahead of them.

⁴² In babies affected with cystic fibrosis, trypsinogen is not converted to trypsin as it normally would be, leaving levels of the precursor to build-up more than usual.

Despite the emotional distress caused in these false-positive cases, it is generally accepted that the beneficence of the program in terms of improved clinical outcomes for babies with CF “[outweighs] the drawback of unwanted carrier detection” (Wilcken, Wiley, Sherry, & Bayliss, 1995, p.965). Although generated incidentally through the program, the “apparent consensus” has been that this genetic information “cannot be withheld from parents” (Miller, Robert, & Hays, 2009, p.210). However, Miller *et al.* have recently called for this “consensus” to be questioned, claiming that “the disposition of carrier results generates competing moral infringements: to withhold information or require its possession” (2009, p.210). It is their view that the automatic disclosure of these results “reflects a bad marriage between clinical bioethics and public health praxis” (2009, p.211).

A CALL TO RESEARCH!

Given the unique features of genetic testing, it is necessarily crucial that the psychosocial welfare of those considering genetic testing is safeguarded by excellent genetic counselling that does indeed attend to their emotional and psychological needs. Despite the importance of the counselling component of ‘genetic counselling’, it has been reported that clinical geneticists deal with this part of the process with “less confidence and less professionalism than when they deal with the more technical and impersonal aspects of their work” (Clarke, 1997d, p.179-180). Consistent with this, clinical geneticists report feeling that they lack training in the skills necessary to achieve all of their own objectives in a genetic counselling session, particularly those required to assess people psychologically and to respond to emotionality (Michie *et al.*, 1999). Nancy Wexler asserted that helping their clients to “[learn] to cope with what is known” requires that a genetic counsellor be “sensitized to detect the subtle psychological effects that the state of genetic risk is likely to produce” (1979, p.200). In saying this, she declared it her firm conviction that:

although individuals vary widely in their reactions to threat, there are still communalities (*sic*) of concern which cut across individual

differences and these can be taught to counselors. In knowing approximately what to expect, the counselor is better able to listen, anticipate, probe, assure, and console (1979, p.200).

In previous research examining psychological reactions to risk factor testing, emotional (how people feel), cognitive (how people think), and behavioural (what people do) outcomes have been assessed (C. Shaw, Abrams, & Marteau, 1999). Research has shown that perceptions of situations as being stressful not only differ markedly between individuals but also vary over time for an individual. Past psychological approaches to understanding this variability have focussed on the role of cognitive processes, including appraisals, expectations of efficacy and perceived control, and causal attributions (Aronson, Wilson, & Akert, 2007; Bandura, 1986; Croyle, 1995; Lazarus & Folkman, 1984). The kind of knowledge that Nancy Wexler (1979) called for requires a depth of understanding that only an interpretive study could provide – one that does not unthinkingly accede to the discourse of science and technology. Thus, it is imperative that pre- and post-test counselling practised today and in the future is **informed** by the experiential expertise of those who have been, or are still, living with a genetic threat.

THE PURPOSE OF THIS STUDY

The broad aim of this thesis is to make an ethical and knowledge-based contribution to the debate about the genetic testing of children. My research will also provide health professionals and genetic counsellors with an insight into the experiences of 'carrier families' – specifically, what it is like to parent (and to be-the-parent-of) a child who has been identified as carrying, or as at high risk of carrying, a mutation in the cystic fibrosis gene. The study is an exploratory one, using an approach based on existential philosophy to investigate and bring to light the experiences of these parents, and it is hoped this increased understanding will impact positively on child and family well-being in families who come into contact with genetic counselling services.

Those who view life as “inherently marginal and ambiguous” urge researchers to “move away from studying abstractions and get at the particular, the detailed and the experiential, that is, ‘concrete human experience’” (Minichiello, Aroni, Timewell, & Alexander, 1995, p.110-111). They reason that this “[allows] the researcher to grasp the ambiguities and inevitability of different perspectives, particularly those of marginal individuals and groups” (Minichiello et al., 1995, p.111). The approach to this study is in line with “[m]uch of the recent discussion within biomedical ethics [which] does move away from abstract, formal principles, stressing instead individual rights, particular contexts, and specific needs” (Diprose, 2005, p.239). Much of the discourse of genetic counselling is still centred on abstract ethical principles, as was discussed earlier (see section: Ethical Issues Facing Genetic Counsellors: Dealing With Past, Present and Future).

With each genetic counselling session being

a unique configuration of personal experience, of familial and peer pressures (or lack of them), of religious and spiritual beliefs (or lack of them), of connections of specific histories to the genetic disease (or lack of them), and, of course, the social and cultural meanings attached to each (Duster, 1990, p.138),

it would be impossible, and effectively without meaning in the ‘real world’, to try to tease out how each factor impacts on the counsellee’s feelings and decisions in relation to their new genetic knowledge. An existential approach is all-embracing; accepting that all of these things are a part of what it means to be human. As will be delineated in this thesis, the individual, having been ‘thrown’⁴³ into the world (i.e., into a particular body, family, community, culture that was not of their choosing), exists in-situation⁴⁴ (i.e., they cannot detach themselves from what they have already experienced in life); has a framework of interpretation and belief⁴⁵ through which they receive and process and then act upon information; is indebted

⁴³ To be discussed in Chapter Three (see section: *Freedom and Choice*).

⁴⁴ To be discussed in Chapter Three (see section: *Ambiguity*).

⁴⁵ To be discussed in Chapter Three (see section: *Freedom and Choice*).

to their community⁴⁶; and despite their freedom, is restricted in their choices by the responsibility they must show toward themselves and toward others⁴⁷.

The purpose of this thesis is to provide an understanding of how the experiences of the parents in this study are shaped by what it means to be human. This will provide health professionals and genetic counsellors with a means of reflecting on the way they provide genetic information to families in whom a recessive disease gene is uncovered, whatever the context or situation.

THE STRUCTURE OF THIS THESIS

This thesis is presented in eight chapters. The first chapter has provided an overview of the debate about the ethical issues surrounding genetic testing and has highlighted, in particular, those in relation to the carrier testing of children. This has set the context for this study, which uses an existential approach to examine the experiences of parents who know that their child is, or could be, a carrier of a CF gene mutation. It highlights the significance of this work as a novel approach to looking at the ethical issues involved in the genetic testing of children. A justification of my decision to adopt a methodology steeped in Existentialism is presented in Chapter Two. A summary of its evolution as a philosophical movement, along with the central concerns of its protagonists are also outlined in this chapter. In Chapter Three, I delineate a conceptual framework that is based on my understanding of existential philosophy. This would then become the 'method' used to analyse the data from the interviews in this study. In Chapter Four, consideration is given to the study design and qualitative research methods that have been used. This chapter includes a discussion of the in-depth interviewing method, describes the procedures used in analysing the data and provides the biographies of the study participants. It ends with my development of an existentialist 'ethic', which I use as a tool to judge the decisions and values that are

⁴⁶ To be discussed in Chapter Three (see section: *Freedom and Responsibility*).

⁴⁷ To be discussed in Chapter Three (see sections: *Freedom and Responsibility* and *Freedom and Choice*).

enacted in the genetic counselling/testing context, as exemplified by the data in this study. The findings of this study have been arranged into three broad areas for analysis and discussion: Knowing Genetic Information, Transcending Genetic Situation and Evading Genetic Threat. These constitute Chapters Five, Six and Seven in this thesis. Finally, in Chapter Eight I highlight the ethical and knowledge-based contribution this study makes to the debate about carrier testing of children, and genetic testing more generally, and suggest possible responses to this research in terms of the practice of genetic counselling and education of the expert ethical professional.

CHAPTER TWO: PHILOSOPHY AND METHODOLOGY

INTRODUCTION

At the end of a four-year Honours degree in Science at university, I found myself trained to be a laboratory-based geneticist – more fondly known as a ‘lab rat’! I was totally immersed in the positivist paradigm and, of more concern to me now, totally oblivious to the fact that I was. Despite a university education in science, I had been taught neither to reflect on the nature of knowledge nor on the question of what can be known nor on the philosophy underpinning the scientific method. Hence, I found myself an embarrassed latecomer to the debate around quantitative versus qualitative research methods, let alone the specific world views that underpin the different paradigmatic approaches to health research.

Some would consider that I use strong language to discuss my rejection of the use of quantitative methods in this study. Quite simply, this has arisen because I set out wanting to understand my participants’ experiences but found myself initially in a research environment where the presumption was that I would, of course, want to achieve this ‘understanding’ through the use of quantitative psychological measures – for what other method could possibly be used to obtain data that could be relied upon; that would have any meaning?! For this reason, I did contemplate a quantitative approach to this study in the early days. I looked at various psychological models⁴⁸ and research tools (measures and surveys)⁴⁹, all the while

⁴⁸ The ‘appraisal of threat’ literature led me to the work on stress, appraisal and coping by Lazarus and Folkman (1984). I pondered using this and the ‘perceived threat’ construct (combines measures of perceived susceptibility and severity) from the Health Belief Model as a theoretical framework for my research. The Health Belief Model is a framework for analysing beliefs that motivate health behaviours and is well established as a model for understanding health behaviour decisions (Strecher & Rosenstock, 1997). I considered conceptualising carrier status as a threat to one’s reproductive future and as a possible threat to one’s children’s health, and saw that self-concept and coping style could be proposed as major determinants of the individual’s response to this stressor.

⁴⁹ A range of approaches to this problem were examined, including previously published questionnaires about perceptions of health and risk perceptions, parental reports of the child’s health, and various methods to quantify the significance of carrier status as compared with other potential stressors experienced in childhood. Examples of specific research tools include the Cognitive Appraisal of Health Scale that was developed to measure multiple dimensions of primary and secondary appraisals associated with health-related events (T. A. Kessler, 1998), the Impact of Event Scale (Horowitz, Wilner, & Alvarez, 1979) for use as a measure of the levels of stress which parents recalled feeling upon discovery of their child’s carrier status and also the Social

taking note of the discourse⁵⁰ that was being used to 'objectively' measure and 'average out' people's experiences, eliminating any space in which a true-to-life story – so vitally important to this enterprise – could be told. The dimensions of the picture these measures and surveys can paint are set in advance, and limiting. It was clear they would not provide a canvas with room for exploration of the experience and would not allow its complexities to emerge.

In the end, it was a case of pointing out the obvious in making the observation that just because people are not showing clinical, 'measurable' signs of depression and anxiety does not mean they do not have real concerns and issues that need to be understood and addressed. Furthermore, each individual's story is highly contextualised – their reactions to genetic threat cannot be conceptualised as merely the product of the disease characteristics (e.g., mode of inheritance + age at onset of symptoms + perceived burden + type of symptoms, *etc.*) and the individual's personal attributes (e.g., socioeconomic status + gender + self-esteem + sense of self-efficacy, *etc.*). And nor should they be if one is to uphold "the dignity of man" in the way that Sartre speaks of it when discussing the subjectivity of the individual:

[T]here is such a truth which is simple, easily attained and within the reach of everybody; it consists in one's immediate sense of one's self.

[...] **[T]his theory alone is compatible with the dignity of man**, it is the only one which does not make man into an object. All kinds of materialism lead one to treat every man including oneself as an object—that is, as a set of pre-determined reactions, in no way different from the patterns of qualities and phenomena which constitute a table, or a chair or a stone. [The existentialists'] aim is precisely to establish the human kingdom as a pattern of values in distinction from the material world. [emphasis added] (Sartre, 1975, p.361)

Readjustment Rating Questionnaire (Holmes & Rahe, 1967) which has been modified to apply to children (Coddington, 1972a, 1972b).

⁵⁰ The discourse concerned itself with 'cognitive processes', 'psychosocial adjustment', 'correlates of adjustment', 'health status', 'health outcomes' and the like.

In conclusion, a quantitative research approach to this inquiry was investigated but for sound epistemological reasons was discounted.

BACKGROUND

A long time ago, or so it seems now, I gathered from my reading that quantitative research largely focussed on predicting and controlling outcomes while qualitative research focussed on describing and understanding experiences.

It was at this moment that my confidence in proclaiming that I definitely wanted to take a qualitative approach to my research increased. For not only did this surge of clarity signal to me that a quantitative approach could not answer my question (and was therefore a scientifically unsound way to go), it gave me a way of explaining to other people why I had chosen this path at a time when my knowledge of qualitative methodologies and their theoretical underpinnings was scant (when my own lack of understanding would have meant that I could not do justice to its merits – and indeed, would have made it sound most unscholarly). It empowered me and enabled me to take a decisive stance about what I wanted to achieve in my project, how I wished to be challenged, where I wanted to sit-in-the-world, and inevitably how I desired others to view me as a researcher.

This method appealed to my personal, ethical, analytical, philosophical, æsthetic and literary sensibilities.

FINDING MY OWN VOICE...

At the time, I felt I had discovered the methodology I wanted to use by following the way I **intuitively** felt the study should be done. To me, it seemed only common sense that if I wanted to understand what was going on with these parents – who had received and lived with the information that their children are carriers of cystic fibrosis – that I should ask them. I looked at various health research texts to gain an understanding of the alternative methods available to answer my particular

research question. In the case of this study it seemed obvious that an in-depth interviewing method would be more respectful of participants' **experiential expertise**, and their own personal and cultural situations that give meaning to the experience for them⁵¹, than giving them a questionnaire with a long arduous, often context-less list of questions that would inevitably belittle the richness of their lived experiences. Looking back, although I was naïve to it at the time, I was actually making a conscious epistemological⁵² choice about the relative value of the kinds of knowledge I could generate in response to my research question.

While many would view qualitative research as a 'soft' way to generate data that gives an insight into the human condition, particularly when compared with the positivist procedures that churn out 'hard' facts, I saw it as a much tougher endeavour – one which requires from the researcher an intimate knowledge and understanding of the data, rather than a reliance upon abstract concepts, models and statistical formulae that apparently 'do their thinking' quite separately from the researcher. In this same vein, Max van Manen has previously described phenomenological research as

a being-given-over to some quest, a true task, a deep questioning of something that restores an original sense of what it means to be a thinker, a researcher, a theorist (1990, p.31).

Qualitative methods are hard work – they challenge the researcher to bring intuition and insight, scholarship and tenacity to the texts which capture a snapshot of the reflected-upon experiences as told by their participants.

...AND LOCATING IT WITHIN A DISCOURSE

Choosing a paradigm is really about choosing a discourse within which you wish your voice to be heard. It is about adopting a world view – choosing a way of being-

⁵¹ Irena Madjar points out that these "situated meanings" must be taken into account if particular human experiences are to be understood and described faithfully (1991, p.50).

⁵² Epistemology is "the study of the philosophical problems in concepts of knowledge and truth" (Collins, 1997, p.198).

in-the-world – and then declaring this to the world. It is saying this is the research question I had and this was the most socially responsible way I could see of going about answering it; this was the way I chose to view the participants in my study; this was the way I wished to present myself to them; this was the sort of knowledge I wished to generate with them; these are the sorts of beliefs, suppositions and assumptions that I make in my everyday lived-reality; and furthermore, that this is the way I would wish to be approached if I was to be a participant in this study; this is the way I would wish to be perceived if I was in need of health care. I needed to ask myself: “For whom am I doing this research?”, “Can I justify asking participants for their time and trust if I do not wish to listen to what they wish to tell me?” and “How do I know what is important to them if I do not give them a voice?”. My being consumed with these questions led to another: how had I come to find myself entirely located within the dominant, scientific, positivistic discourse without consciously seeking a position there? Reflecting on this, of course, I realise that our society, in particular the research community, privileges this discourse over interpretive and critical ones; that I was ‘educated’ into it – it is the default existence (at least for now) or as Heidegger may have put it, the “undifferentiated” existence (Lemay & Pitts, 1994, p.54). So finding another paradigm is hard work. Stepping out of the dominant discourse is a challenge, not just a professional and intellectual challenge but a personal challenge – reason enough, I think, to believe that those who make this leap do so out of a fervent belief in its worth.

I had found phenomenology.

THE LURE OF PHENOMENOLOGY

It is widely accepted that phenomenology as a research methodology is couched within the Interpretive Paradigm – that is, it has been aligned with approaches to human inquiry that are considered to be interpretivist or constructivist. Interpretivism emerged amid critiques of scientism and positivism in the social sciences; being an approach that recognises the uniqueness of human inquiry and rejects the notion that the aims and methods used in pursuit of scientific

explanation in the natural sciences should be equally embraced within the social sciences (Schwandt, 1998, p.223). One of the fundamental assumptions underlying interpretive research is that

we need to know what people think in order to understand why they behave in the ways that they do. This, in turn, is predicated on the belief that people act in the ways that they do because of the way in which they define the situation as they see it or believe it to be. That is, they interpret the facts as they see them. (Minichiello et al., 1995, p.69)

This kind of research allows us to understand “people’s experience of social reality through their routinely constructed interpretations of it” (Minichiello et al., 1995, p.69).

Notwithstanding my satisfaction at discovering this ‘new’ methodology, it was later with some caution that I continued down this research path. And it was with some trepidation that I tried to describe exactly what the phenomenology I believed I was being informed by actually was. My initial contact with phenomenology was via nursing texts about qualitative research (e.g., Streubert & Carpenter, 1995) and studies about illness and nursing practice that had used this methodology (Madjar & Walton, 1999). Their descriptions of what phenomenology was and what it could do were alluring and inspiring. For someone who felt confined within a quantitative research world, the promise of qualitative inquiry and especially the literary style of the phenomenological text was a welcome escape and a heartening affirmation that there was a whole research world where people were roughly on the ‘same wavelength’ as me. But little did I then know about the mishmash of interpretations of, and varying degrees of reliance upon, the philosophical underpinnings of phenomenology by those who claim to conduct phenomenological research (Crotty, 1996). I found the emerging claim that nursing theorists were largely misinterpreting Husserlian and Heideggerian phenomenology concerning, and credible (Paley, 1997, 1998).

While the initial simplistic understanding I naïvely gained from nursing texts about phenomenology indicated that it was a method for those interested in understanding subjective experience (this of course resonating with my wish to look at ‘what it is like for parents who know their children are carriers of cystic fibrosis’) – and indeed van Manen’s *Researching Lived Experience* (1990) provided an illuminating and highly-readable exposition of how this kind of phenomenological text could be produced in an exquisite way – as I came to understand a little more of Husserl’s philosophisings, and then understand these with more confidence via Crotty (1996), I discovered that ‘pure’ phenomenological research has a “note of *objectivity* about it” and is, in fact, “an exercise in *critique*” (Crotty, 1998, p.82-83). Phenomenology is “in search of objects of experience [and the essences which constitute them] rather than being content with a description of the experiencing subject” (Crotty, 1998, p.83). It invites us to “engage with phenomena in our world and make sense of them directly and immediately”; to “lay aside, as best we can, the prevailing understandings of those phenomena and revisit our immediate experience of them” – that is, “before we [started] thinking about them, interpreting them or attributing any meaning to them” (Crotty, 1998, p.78-79).

With this in mind, I realised I was unable to define, in phenomenological terms, the nature of the beast I had chosen to research. Would ‘carriership’ actually be considered a phenomenon, ‘object’⁵³, experience, or ‘thing in itself’ to the ‘expert’ phenomenologist? How could a state with a perfectly infallible and objective definition be mined for its constituting essences⁵⁴? And if carriership is not a phenomenon constituted by essences, then how could it be the subject of phenomenological research? I asked, not without some cynicism, and perhaps with some arrogance, whether a solid understanding of the seminal phenomenological texts (written for philosophers more so than researchers) would truly add to my analysis in any significant way over and above what a keen, interpretive, insightful,

⁵³ Husserl’s use of the word ‘object’ refers to “things in the external world, facts, concepts, pains, the data of consciousness, dream images, essences...anything” (Paley, 1997, p.190).

⁵⁴ Phenomenological research is the study of essences. Crotty draws on his understanding of Husserl’s and Merleau-Ponty’s work to define an ‘essence’ as “the very nature of a phenomenon, [...] that which makes a some-‘thing’ what it *is*—and without which it could not be what it is” (1996, p.10).

common sense would. That is, I was anxious that I not present an exegesis of philosophical tenets in the methodology chapter that would then not flow through to the data analysis chapters. I certainly did not want to write about phenomenological philosophy merely in order to prove myself as a research scholar if I felt that, in the final analysis, my understanding of this had done little to enrich the depth or scholarliness of my actual analysis chapters. After all, I asked myself, would Husserl or Heidegger have recommended their philosophical texts to a researcher who was wishing to go about the task of analysing a large amount of qualitative interview data? How would they have suggested we 'concretise' their philosophy into method?

As I was immersed in the world of nursing and midwifery research, it became increasingly clear that there was much contention among nursing and midwifery academics about the value of interpretive research, that is, about the value of the kind of knowledge it could produce. In conversations 'around the water cooler', a researcher avowedly devoted to the Critical Paradigm would vehemently deny that phenomenology is critical or that my data could be subject to a critical analysis. When I put forward Crotty's assertion that "[t]he phenomenology of the phenomenological movement is a thoroughly critical methodology" (Crotty, 1996, p.4), another would ask me if I claimed to be empowering my participants – because I could not say I was using a critical methodology if I was not doing this. I found myself arguing that the notion that I could empower and enlighten my participants merely through interview (without the corresponding analysis, critique and process of change that comes with action research methods) was condescending and certainly not the province of my research. Rather, I said I hoped to be critical by offering an interpretive exposé of our collective construction of carrier status as a society (as reflected in the individual narratives of my participants) and that, by doing this, I just may challenge prevailing conceptions and our received notions and taken-for-granted assumptions about what carriership means within families in the sociohistorical context that is Australia today. After all, "in order to see the world and grasp it as paradoxical, we must break with our familiar acceptance of it" (Merleau-Ponty, 1962, p.xiv).

For me, a critical methodology is one in which lies the potential to enact change – whether it be for individuals, within institutions or across societies – and surely a rethinking of one’s acceptance of ‘the way things are’ and an empathic understanding of the situation of the ‘Other’ are stepping-stones on the way to change? That is not to say I am declaring such transformative power will lie within the pages of my thesis alone, rather that one should not be so quick to dismiss the potential of a piece of research to generate critical findings on the basis that it aligns itself with a methodology that does not lie within what is widely accepted as the Critical Paradigm.

FROM PHENOMENOLOGY TO EXISTENTIALISM

Something changed. I was no longer satisfied that what I was **doing** was phenomenology at all. I was **talking** to people about one aspect of their lives but I could not even say it was the “subjectivist” and “uncritical”⁵⁵ brand of phenomenology – that I was ‘researching lived experience’ – as a good part of the interviews focussed on the parents’ outlook on the future for their children, not just those experiences that had already been lived. I was talking to my participants because it would have been futile to observe, as an ethnographer would, the parents of carriers in their ‘natural habitat’ (i.e., in their “real-life context”) engaged in “mundane and practical human activities” – existing in their “everydayness”⁵⁶ – and then to try to seek, from amidst my observations, the nature of the ‘carrier’ phenomenon and the essences which constitute it. After all, what would I have seen? You **cannot see** a person going about their day as the parent of a carrier as

⁵⁵ Crotty uses these words to describe the brand of phenomenology that has come to dominate in nursing research. By ‘subjectivist’ he means “in the sense of being in search of people’s subjective experience”; by ‘uncritical’ he means in its inability to “capture new or fuller meanings”, which may mean “oppression, exploitation and unfreedom are permitted to persist without question” (1998, p.83/85).

⁵⁶ These phrases are borrowed from Crotty in his discussion of Heidegger’s work. He says that Heidegger “starts his inquiry from the living human being in that human being’s real-life context” (1996, p.84). By engaging in a “phenomenology of everydayness”, Heidegger’s aim is “to reveal the primary understanding of the world which he sees as informing all our day-to-day interpretations” (Crotty, 1996, p.84). To this end, *Being and Time* (Heidegger, 1996) was “filled with examples drawn from mundane and practical human activities” (Crotty, 1996, p.84).

you could perhaps watch a fire-fighter or a teacher at work and look to the defining essences of what it is to be in those particular occupations. Directly experiencing their social world would have revealed nothing to me about the nature of this dubious phenomenon. I say 'dubious' because the idea that carriership is an object (in the phenomenological sense of the word) constituted by essences was no longer plausible for me.

Any geneticist can give a definition of what a carrier is: a healthy person with only one faulty copy of a particular recessive disease gene. One cannot talk about carriership as some predefined, universal, unexperienced, yet-to-be-reflected-upon state – as a Husserlian phenomenologist would be inclined to do – without reducing it to a meaningless concept. Nietzsche's rebuttal of Kant is important here. Kant postulated that humans have particular ways of interpreting (time, space, etc.) that help to filter our experience of particular objects and phenomena; that there are 'ideas' in the mind that resemble what is found in the external world around us (Flynn, 2006, p.71); that consciousness "finds in things only what it has put into them" (Merleau-Ponty, 1962, p.439). He believed that we all have the same 'filters' and that therefore universal human knowledge can be generated. This theory was opposed by Nietzsche on the basis that it assumed "that some sort of universal 'truth' existed to be discovered" (Lemay & Pitts, 1994, p.21). The assumption that there are universal human truths that can be derived from the universal categories of interpretation in our minds does not take into account the all-pervasive effect of culture (at a macro and micro level) on the way individual and collective human minds come to **learn to interpret** over their lifetime.

So in investigating the experience of these parents, a Husserlian phenomenologist would say that culture constantly blocks our access to this object called 'carrier', yet, in reality, it is culture that has brought forth the very existence of this thing we call 'carrier'. This phenomenon would not exist without culture so to seek to find something akin to the Platonic Form of Carrier (i.e., the universal essences⁵⁷ of the

⁵⁷ In Plato's Theory of Ideas, "essence is the grasp of the very nature of something, of which any particular instance is only an imperfect example or imitation" (van Manen, 1990, p.177).

phenomenon of 'carriership') devoid of a human interpretive lens is an impossible task – in other words, the eidetic reduction⁵⁸ is not possible here. Thus my reason for saying that the object called 'carrier' does not exist in a form which can be the subject of a phenomenological investigation, but the existential states of being-a-carrier and parenting-a-carrier are open to an investigative foray. And, by understanding the way enculturated beings experience being-a-carrier and parenting-a-carrier, we gain access to the 'phenomenon' of carriership that exists only because humans have socially constructed it.

So if initially we think about the 'whatness' of carrier status (as is the wont of those influenced by phenomenology), we soon realise that we must not focus on this thing-in-itself called 'carrier' but rather on **being-a-carrier** and **parenting-a-carrier**. In other words, we need to seek to understand not what a carrier is but what the human experience of being-a-carrier is; what this means to the individual who is said to 'carry' a disease gene. We need to uncover the situated knowledge that is derived from the experience of parenting-a-carrier and look to how the human mind filters this experience in its own unique way. We see that it is now clear an existential approach to this study was needed, rather than a phenomenological one. This required that I look to existential philosophy in order to develop an analytical framework which could be used to interpret the data generated from my interviews with the parents participating in this study. The rest of this chapter is devoted to an overview of the philosophy and the central concerns of its protagonists.

EXISTENTIALISM...IN THIS THESIS

It is beyond the scope of this thesis to re-interpret existentialist texts or to re-invent an exegesis of Existentialism of which there are a number (for examples, see:

⁵⁸ It is from the Greek word *eidōs* that the adjective 'eidetic' is derived. *Eidōs* was used by Plato as an alternative term for his concept of the Idea or Form before Husserl adopted the word to designate a universal essence (Crotty, 1996, p.32). The 'eidetic reduction' is a phenomenological device, which according to Husserl, permits us to come to an understanding of the essential structure of something. In order to achieve this, "one needs to see past or through the particularity of lived experience toward the universal, essence or *eidōs* that lies on the other side of the concreteness of lived meaning" (van Manen, 1990, p.185).

Charlesworth, 1975; Cooper, 1990; Flynn, 2006; Olson, 1961). Even Thomas Flynn describes Sartre's masterwork, *Being and Nothingness*, as a "long and difficult book" – "not exactly a bestseller" – and he concedes that "like Darwin's *The Origin of Species*, it was more often cited than read" (2006, p.46). Accordingly, I have heavily relied upon Charlesworth in order to present the evolution of Existentialism as a philosophical movement and Cooper has provided an excellent basis for understanding Existentialism as a collection of concepts that occupy the thoughts of existentialist philosophers. I have presented here that which may be understood by the non-philosopher.

THE HISTORICAL AND INTELLECTUAL ANTECEDENTS OF EXISTENTIALISM

The philosophy and the plays and the politics of Jean-Paul Sartre, the novels of Albert Camus, the writings of Simone de Beauvoir, the dark metaphysical utterances of Martin Heidegger, the autobiographical philosophizings of Gabriel Marcel – all were the expression of a pervasive intellectual mood in post-war Europe; and they gave a dramatic voice to the bleak and disenchanted spirit of that age (Charlesworth, 1975, p.1-2).

Existentialism is an interdisciplinary movement that finds expression in philosophy, literature and psychotherapy. It has been more curiously described as "that complex philosophical-literary-political-psychoanalytical-religious and anti-religious movement of the 1940s and 1950s which later came to be known as Existentialism" (Charlesworth, 1975, p.1). Existentialism became a great intellectual fashion and vogue in Europe, and especially in Paris, at this time, largely due to the timeliness of the works of the French philosophers and novelists, Jean-Paul Sartre (1905-1980), Simone de Beauvoir (1908-1986) and Albert Camus (1913-1960), and the celebrity accorded to them. The mood amongst the intellectuals who frequented the Left-

Bank Parisian cafes was “one of enthusiasm, creativity, anguished self-analysis, and freedom – always freedom” (Flynn, 2006, p.9).

After the two world wars were over,

everyone was ready for a philosophy that could nod to the irrational elements in life; hence, perhaps the immense popularity of both psychoanalysis and existentialism after the abattoir of the twentieth century (Marino, 2004, p.xiv).

Simone de Beauvoir herself explained that

[t]he popularity of Existentialism just after the war and Sartre’s incentive to think along these lines, are both phenomena which can be explained by reference to the historical situation at the time. Both we and the people who read our works felt the need of an ethical system of norms and Existentialism served to reconcile history and ethics (interview with Pierre Vicary cited in Charlesworth, 1975, p.6).

According to de Beauvoir, the first existentialist philosophers were Søren Kierkegaard (1813-1855), Martin Heidegger (1889-1976) and Karl Jaspers (1883-1969), after whom came “the Christian Existentialism of Gabriel Marcel” (1889-1973) and “the atheistic Existentialism of Sartre” (interview with Pierre Vicary cited in Charlesworth, 1975, p.5)⁵⁹.

Existentialism is difficult to define, partly because “almost everyone who was labeled an existentialist went to great lengths to deny that he or she was an existentialist” (Marino, 2004, p.xiii). Influenced by Kierkegaard, Jaspers (existential philosopher and psychiatrist) was using the term *‘Existenzphilosophie’*⁶⁰ after the First World War but it is generally accepted that the word ‘existentialism’ did not come into currency until the mid-1940s (Marino, 2004, p.xii). Simone de Beauvoir believed Existentialism was born in 1945 when, she said, she and Sartre “took the

⁵⁹ Interviews are seen as a valid reference source in the interpretive paradigm where the criterion of what ‘counts’ as knowledge is broad. This is in opposition, say, to the field of psychology in which this would not traditionally be accepted as a valid source of understanding because there has been no opportunity for the text to be peer-reviewed, a prerequisite to it being recognised as validated knowledge; as ‘true’.

⁶⁰ The English translation is ‘Existence philosophy’.

epithet that everyone used for us and used it for our own purposes" (De Beauvoir, 1965, p.38). Previously, Sartre had refused to allow Gabriel Marcel to apply the adjective, 'existentialist', to his work, and is quoted by de Beauvoir as saying, "My philosophy is a philosophy of existence; I don't even know what Existentialism is" (De Beauvoir, 1965, p.38). However, just after the end of World War II, Sartre gave a public lecture entitled, *L'Existentialisme est un Humanisme*⁶¹, which "served as a quasi-manifesto for the movement" (Flynn, 2006, p.x). It was apparent by then that Sartre had wholeheartedly accepted the name 'Existentialism' for his and de Beauvoir's philosophy. Despite this, Camus persisted with the assertion that he himself was not an existentialist because, as a novelist, his work was quite different to the philosophy of Sartre and Heidegger. In turn, Heidegger too refuted any suggestion that he was an existentialist, at least as Sartre conceived of it (Lemay & Pitts, 1994, p.68-69; Marino, 2004, p.xiii).

Once Existentialism took hold in America in the late 1940s and 1950s, scholars began tracing back from Sartre to Heidegger to Friedrich Nietzsche (1844-1900) and Kierkegaard to determine from whence the elements of Existentialism arose. Some claim that the lineage of existentialists can be traced back to St. Augustine of Hippo (AD 354-430), while others classify St. Thomas Aquinas (c. 1225-1274), William Shakespeare (1564-1616) and Blaise Pascal (1623-1662) as early existentialists (Marino, 2004, p.xv). Kierkegaard, the "Danish theologian concerned to rescue Christianity from legalism and institutionalism" (Charlesworth, 1975, p.10), is widely referred to as the 'Father of Existentialism'. However, Kierkegaard, the German thinker Nietzsche and the German professor of philosophy Edmund Husserl (1859-1938) have all been touted as the ancestors of Existentialism (Charlesworth, 1975, p.10).

More recently, the existential movement has been described by the philosopher Charles Guignon (who was drawing on the work of Max Weber) as a response to the disenchantment of the world; to the sense that the history and social structure of

⁶¹ *Existentialism is a Humanism* is Philip Mairet's translation of the lecture (Sartre, 1975, p.345-369).

the world are not God-sanctioned (Marino, 2004, p.xiv). Indeed, it has been said that the roots of the existential approach emerged during the Enlightenment “when the faith of people turned in varying degrees from God to reason and humanity itself” (Marino, 2004, p.xiv).

John Carroll discussed the historical and intellectual antecedents of Existentialism in a radio interview with Max Charlesworth (documented in Charlesworth, 1975)⁶². He described Existentialism as part of a tradition which has its seed in Renaissance humanism:

With the Renaissance there was introduced the notion that man, and maybe the individual man, has some sort of primacy and centrality in the universe. What is central is not God, and it's not some divine hierarchy deriving from God, or even some social hierarchy with analogies in divine hierarchy such as the medieval view would have had. It is not these types of order which are primary. Rather, the individual has central and unique power (Charlesworth, 1975, p.10).

In the course of the interview, Carroll moved on to show how these roots of existentialist thought were then nurtured by the fertile environment provided by Calvinism. He said:

[T]he Calvinist belief is that a man's soul, the quality of his soul, his virtue, is a matter to be decided, not by his fellow citizens, but by himself, the lonely individual in relationship to his god. [...] [T]his brought with it, not only an undermining of community values, but also an emphasis on introspection, an emphasis on privacy, on the inner man, an extreme rationality about psychology and about theology, and [...] an emphasis on solitude (Charlesworth, 1975, p.10).

He followed with an explanation of the way in which these existentialist offshoots were then nurtured in the folds of the Romantic Movement. He highlighted this by encapsulating the movement as one that now

⁶² Max Charlesworth's radio programme featuring the interview with Dr John Carroll of LaTrobe University in Melbourne was called *The Existentialists*, and was broadcast on the Australian Broadcasting Commission's Radio 2 early in 1975. The text of the programme was published as Part 1 of a book later that same year and that is the reference given here.

[portrayed] the society or the collectivity as something which is completely repressive of the individual, standing over against the individual, and at the same time [placed] a very high value on the individual's only feelings and on his own experience of life, on the whole complex of inner impulses and inner sensations which come to an individual in his life (Charlesworth, 1975, p.11).

Evidently, most existentialist philosophers are individualists.

For Carroll, Existentialism's emphasis on the primacy of the ego or the self, "implied in the hostility to role-playing and social roles which one finds in Heidegger and Sartre, is a very romantic notion of the individual standing over against society, a romantic notion in which the individual is the centre for human value" (Charlesworth, 1975, p.11). Indeed, the existentialist's attention is always focussed at the level of personal meaning rather than general theory. For example, in reflecting upon what it means to die, Kierkegaard, and then Heidegger, "try to fathom the individual meaning of our mortality" whereas other philosophers may have tried to formulate a general theory of personal identity or even a definition of death (Marino, 2004, p.xii). Such is Existentialism's emphasis on the individual, some existential philosophers have insisted on working from the first-person perspective (Marino, 2004, p.ix).

HUSSERLIAN PHENOMENOLOGY

While Husserlian phenomenology was not employed as the research methodology for this study, a temporary detour must be made back to Husserl here because the phenomenological method he developed was "adopted in one form or another" by the existentialists of his time (Flynn, 2006, p.17). Many phenomenologists were not existentialists (Flynn, 2006, p.17) but it was the phenomenological work of Husserl that gave rise to Existentialism as a purely philosophical movement (Marino, 2004, p.xv).

Husserl's emphasis was on taking philosophy back to the analysis of concrete experience. Husserlian phenomenology "brackets the question of whether or not

our experience points to anything beyond itself and sticks to the analysis of experience itself" (Marino, 2004, p.xv). In the words of Camus in *The Myth of Sisyphus*, "phenomenology declines to explain the world, it wants to be merely a description of actual experience" (2004a, p.471).

Husserl articulated an approach which he believed would enable philosophy to become a "body of rigorous, necessarily certain, universal, self-evident, self-justifying, truths" (Charlesworth, 1975, p.21):

This method, which he called phenomenology, consisted in the philosopher putting himself into a position from where he could intuit and describe things as they appeared to him directly or immediately before he began reflecting or interpreting or placing his constructions on what he experienced. The recovery and description of this primordial contact with the world, this naïve and spontaneous and unreflective experience upon which all our other contacts with the world are built up, is for Husserl the main task of philosophy. It is also a supremely difficult task for we have to purge our minds of all kinds of preconceived ideas and expectations which prevent us from seeing things as they really are (Charlesworth, 1975, p.21).

To do this, he proposed

[m]ultiple processes of phenomenological *reduction*⁶³ [that he believed would] make it possible to *bracket* or disconnect the world's 'taken-for-granted reality', and to gain a state of pure consciousness or *ego*. This consciousness is viewed as a self-contained, self-sufficient entity, existing apart from and continuing beyond the physical world. The disengaged consciousness can then be directed towards a specific *focus*, leading to a dual process of conscious awareness and reflective consciousness (Grbich, 1999, p.168).

⁶³ These processes have been summarised by van Manen (1990, p.185) for the researcher of lived experience; the 'reducer'. He says one needs: 1) "a profound sense of wonder and amazement at the mysteriousness of the belief in the world"; 2) "to overcome one's subjective or private feelings, preferences, inclinations, or expectations that would prevent one from coming to terms with a phenomenon or experience as it is lived through"; 3) "to strip away the theories or scientific conceptions and thematizations which overlay the phenomenon [...] and which prevents one from seeing [it] in a non-abstracting manner"; and 4) "to see past or through the particularity of lived experience toward the universal, essence or *eidōs* that lies on the other side of the concreteness of lived meaning" (1990, p.185).

Since most would reject Husserl's 'brand' of objectivity as impossible to achieve – as stated by Merleau-Ponty, "you cannot 'reduce' the existing 'reducer'" (cited in Flynn, 2006, p.23) – many would then perceive his emphasis on a return to experience as nothing more than a promotion of subjectivism. One may imagine that a description of our primordial experiences would reveal more about our own subjective states than an accurate depiction of the objective world. However, Husserl met this objection by introducing the principle of intentionality, where awareness is inevitably **awareness of** an object.

The theory of 'the Intention', made fashionable by Husserl and the phenomenologists, was an extension of Brentano's⁶⁴ idea that consciousness is always *consciousness of* (Paley, 1997). Charlesworth explains:

All my conscious acts are intentional, that is to say they are not just psychological processes which take place within me in some inner, private, "subjective" world; rather they are all directed outwards to the objective world. To think, for example, is not for some psychological process to take place within the privacy of my psyche; it is rather to be conscious *of* some object outside my psyche. And the same is true of all our conscious acts; they are object-directed or "intentional"; they *ipso facto* involve us in the world outside ourselves (1975, p.21).

In other words, Flynn explains,

it is the very nature of consciousness to aim towards (to 'intend') an other. Even when it is directed towards itself in reflection, consciousness is directed as towards an 'other'. [...] In this context, 'intentional' [...] is a technical term for what is unique about our mental acts: they extend beyond themselves towards an other. (2006, p.17)

In 'the Intention', Camus tells us that

there is no truth, but merely truths. [...] [E]verything has its truth. Consciousness illuminates it by paying attention to it.

⁶⁴ Franz Brentano (1838-1917) was "an outstanding philosopher and psychologist", and Husserl's teacher (Crotty, 1996, p.36).

Consciousness does not form the object of its understanding, it merely focuses, it is the act of attention [...] **Consciousness suspends in experience the objects of its attention.** Through its miracle it isolates them. Henceforth they are beyond all judgments. This is the "intention" that characterizes consciousness. [emphasis added] (Camus, 2004a, p.471)

One of Husserl's main preoccupations was "to understand the relationship between awareness of particular objects (in this unusual sense of the word) *as* particulars, and awareness of them as examples of general concepts" (Paley, 1997, p.190).

THE EXISTENTIALIST CRITIQUE

Husserl's existentialist followers interpreted him in a way which he would have neither understood nor approved. The existentialists "implicitly reject Husserl's ideal of philosophy as a body of necessarily true, self-evident, self-justifying truths" (Charlesworth, 1975, p.21). Heidegger, Sartre, and their French contemporary, Maurice Merleau-Ponty (1908-1961), were unanimous in dismissing Husserl's programme of 'pure' phenomenology as impossible because "[o]ne can neither doubt, nor seriously pretend to doubt the reality of the world" (Cooper, 1990, p.5). Further to this, Merleau-Ponty asserted that consciousness cannot imagine itself bereft of a world for it "always finds itself already at work in the world" (Merleau-Ponty, 1962, p.432).

However, while some ideas central to Husserlian phenomenology were rejected, the existentialist thinkers took some of Husserl's ideas and used them for their own purposes:

they retain his idea that the method of philosophy must be that of the intuitive understanding and the description of those primordial experiences upon which everything else is based; and they also retain his idea of "intentionality" which undercuts the classical separation of the inner, private, subjective world of consciousness from the outer, public, objective world of things and facts (Charlesworth, 1975, p.21).

According to Charlesworth (1975, p.22), Maurice Merleau-Ponty (not strictly thought of as a member of the Existentialist movement, but rather more a phenomenologist) clearly expressed the Husserlian themes of 'intuitive understanding' and 'intentionality' in the preface to his book *The Phenomenology of Perception*:

It is a matter of describing, not of explaining or analysing. Husserl's first directive to phenomenology, in its early stages, to be a 'descriptive psychology', or to return to the 'things themselves', is from the start a forswearing of science. I am not the outcome or the meeting-point of numerous causal agencies which determine my bodily or psychological make-up. I cannot conceive myself as nothing but a bit of the world, a mere object of biological, psychological or sociological investigation. I cannot shut myself up within the realm of science. All my knowledge of the world, even my scientific knowledge, is gained from my own particular point of view, or from some experience of the world without which the symbols of science would be meaningless. The whole universe of science is built upon the world as directly experienced, and if we want to subject science itself to rigorous scrutiny and arrive at a precise assessment of its meaning and scope, we must begin by reawakening the basic experience of the world of which science is the second-order expression. [...] To return to things themselves is to return to that world which precedes knowledge, of which knowledge always *speaks*, and in relation to which every scientific schematization is an abstract and derivative sign-language, as is geography in relation to the countryside in which we have learnt beforehand what a forest, a prairie or a river is (Merleau-Ponty, 1962, p.viii-ix).

The participants in this study will not be conceived "as nothing but a bit of the world" – they are not seen as "mere objects" of this investigation. I recognise that my knowledge of the world and my participants' knowledge of the world are gained from our own respective particular viewpoints but this speaks to multiple 'truths' about the experience of 'carrier families', rather than one. There is no 'thing in-itself' to return to, only what these families make of their situation. And, as distinct from the phenomenology of which Merleau-Ponty speaks, **I do claim to explain and**

analyse my data⁶⁵, not merely describe it, and I use the insights provided by a conceptual framework that I have derived from existential philosophy to do this.

RECONCILING THE CHRISTIAN AND THE ATHEIST EXISTENTIALISTS

For the newcomer to a philosophy so focussed on the human condition and the meaning (or lack thereof) of life, it may seem difficult at first to reconcile the notion that some existential philosophers were atheist, some agnostic and some religious. Charlesworth (1975, p.8) summarised their various positions thus:

Sartre [...] is an atheist, and it is very important for him that God does not exist. Gabriel Marcel, on the other hand, was a devout Catholic; Jaspers a kind of non-sectarian theist; Heidegger an ambiguous atheist; Camus a devout agnostic;

neatly reflecting how radically disparate their beliefs with regards to the existence of a 'higher being' were.

The Father of Existentialism himself was a highly religious man; Søren Kierkegaard was a Lutheran whose authorship dwelt heavily on the question of what it means to be a real and authentic Christian. He eventually came to the conclusion that "believing in Christ involved the attempt to follow Christ in his self-denial, his suffering, and ultimately in his humiliation" and yet, he asserted, "the imitation of Christ was about the furthest thing possible from the mind-set of institutionalized Christianity" (Marino, 2004, p.4). He probably did not regard himself as a philosopher and his aim was never to develop a consistent, systematic philosophical position, rather his whole life was directed towards reforming Christianity (Wardlaw, interview with Charlesworth, 1975, p.13)⁶⁶. For Kierkegaard, reform was necessary because

⁶⁵ However, I do not make the further claim to "prove anything", only to "show some things" (Heidegger n.d. cited in Charlesworth, 1975, p.9). My need to state this will become evident upon a reading of Chapter Three (see section: The Existentialist 'Method' Focuses on Concrete Experience).

⁶⁶ Professor Harry Wardlaw of Ormond College at the University of Melbourne was also interviewed by Max Charlesworth for his radio programme, *The Existentialists*.

[b]eing a Christian has nothing to do with belonging to a sect, or believing a set of doctrines, or performing certain rites; it is rather a matter of taking up a certain basic attitude to life, adopting a certain fundamental "life-style", existing in a certain way (Charlesworth, 1975, p.11).

According to the Dane, Christianity is not a doctrine but rather an "existential communication", for an attentiveness to Christianity cannot be acquired by "reading books or by world-historical surveys, but [only] by immersing oneself deeper in existence" (Charlesworth, 1975, p.11).

Kierkegaard delineated and described the æsthetic, ethical and religious modes of life. He outlined the æsthetic way of life as one "of personal inclination, following out one's own interests and ideals, following one's own star, making of one's life a work of art" and contrasted it with the ethical way of life, which "is actively concerned with the good of others, and it is lived by reference to universal moral principles" (Charlesworth, 1975, p.11-12). The religious way of life transcended both of these, being a life that "go[es] beyond what is required by any principle, being willing to do more than any obligation demands" (Charlesworth, 1975, p.12).

The paradoxical nature of Kierkegaard's influence on later existential thinkers has not gone unnoticed, in that "what was for him a way of reforming Christianity, [was] taken over [...] and used to elaborate a radically secularist, a-religious, view of Man" (Charlesworth, in his interview with Wardlaw, 1975, p.12). Camus, Sartre and Heidegger secularised Kierkegaard's philosophising by promulgating what he held to be true of the specifically religious mode of existence as true of life in general (Charlesworth, 1975, p.12) – that "the single individual is higher than the universal"⁶⁷ (Kierkegaard, 2004a, p.9). In life, having been in the universal, the existentialists believe it is the task of the single individual to isolate themselves as higher than the universal.

⁶⁷ The ethical ('social morality') is the 'universal', and as the universal it applies to everyone at all times (Kierkegaard, 2004a, p.7).

Camus, Sartre and Heidegger were all deeply influenced by Kierkegaard but it was not the religious dimension of his thought which influenced them, rather it was his insistence upon the individual and the 'lived experience'⁶⁸ of the individual (Charlesworth, 1975, p.12). He had proposed a radical and extreme individualism that saw the individual person considered in isolation from history and the social relationships in which they were enmeshed (Charlesworth, 1975, p.14). He had described the anonymous 'public' thus:

[T]he public is [...] an abstract void and vacuum that is all and nothing [...] the most dangerous of all powers and the most meaningless. One may speak to a whole nation in the name of the public, and yet the public is less than one ever so insignificant actual human being. The category "public" is reflection's mirage delusively making the individuals conceited, since everyone can arrogate to himself this mammoth, compared to which the concretions of actuality seem paltry. The public is the fairytale of an age of prudence, leading individuals to fancy themselves greater than kings, but again the public is the cruel abstraction by which individuals will be religiously educated—or be destroyed.

[...] In this state of indolent laxity, more and more individuals will aspire to be nobodies in order to become the public (Kierkegaard, 1978, p.93-94).

It was Kierkegaard's contention that the bankruptcy of a bourgeois society distracts a person from their own individual responsibility (Wardlaw, interview with Charlesworth, 1975, p.14), that is, to have a direct unmediated relationship with God. He believed that the 'public' threatens "a person's 'individual religious isolation', his capacity to enter, alone and unaided, into that personal relationship to God which is the highest human aspiration" (Cooper, 1990, p.112)⁶⁹. According to Flynn, he attacked not only the State Church for doing "[people's] believing for them" but other potent forces of conformity in Copenhagen in his day too – the

⁶⁸ The existentialist senses the need to 'personalise' the most impersonal and objective phenomena in our lives, including space and time (Flynn, 2006, p.5). They view scientific conceptions of space and time as abstractions from the lived experience of existential space and time (Flynn, 2006, p.67). 'Lived space' and 'lived time' are two dimensions of our lived experience. They are notions that describe our subjective experience of space and time; how they exist within, and present themselves to, our consciousness.

⁶⁹ Here, Cooper cites Kierkegaard, *The Present Age*, Harper & Rowe, 1962, p.54.

popular press for doing “[their] thinking for them” and Hegelianism, the prevailing philosophy, for doing “their choosing for them”⁷⁰ (2006, p.25-26).

Charlesworth explains that Camus, Sartre and Heidegger also shared Kierkegaard’s

distrust of abstract speculative thought which objectifies what is essentially subjective and personal, classifies and generalizes what is essentially unique and particular, leads us to think about things and to stand apart from reality instead of immersing ourselves in it and actually living it (1975, p.12).

Indeed, Kierkegaard and the other existentialists were less concerned with epistemological questions, such as the validity of knowing and the problem of subjectivity, than with the question of how people should live and the search for a “Philosophy of Life” (Cowburn, interview with Charlesworth, 1975, p.15)⁷¹.

Kierkegaard’s charge against Hegel was that he had “built his great philosophical system [...] and then lived in a little hole alongside it” (n.d. cited in Charlesworth, 1975, p.9). Whereas much academic philosophy may be seen as far removed from experience and wisdom, the existential movement “[resonates] with the ancient view of philosophy as a way of life, as a guide for the perplexed” (Marino, 2004, p.xi). It is little wonder that Existentialism has been described as “a perspective that articulates and responds to the deepest human needs” (Mooney, in praising ‘Basic Writings of Existentialism’, as cited in Marino, 2004). Existentialist thought can have a profound effect on the lives of its readers. One famous example is that of the U.S. politician, Robert Kennedy, who was an avid reader of Camus’ work and deeply influenced by him – he “memorized him, meditated about him, quoted him and was changed by him” (extract from a memorial article cited in Charlesworth, 1975, p.4).

Aside from Kierkegaard, Friedrich Nietzsche’s influence on the existentialists also ran very deep. In fact, it has been said that the German thinker, seen as “[t]he early

⁷⁰ In saying this, Kierkegaard was referring to his view that Hegelianism “‘mediated’ otherwise individualizing choices in some higher, encompassing viewpoint in a process called ‘dialectic’” (Flynn, 2006, p.25-26). In opposition to this, “existential thinking stresses choice [between alternatives], the ‘either/or’ that involves risk, commitment, and individuation” (Flynn, 2006, p.32).

⁷¹ Father John Cowburn of the Jesuit Theological College in Melbourne was also interviewed by Max Charlesworth for his radio programme, *The Existentialists*.

psychoanalyst of our culture”, was “the key to the development of Existentialism” (Carroll, interview with Charlesworth, 1975, p.19/18). Nietzsche was drawn to the writings of the German philosopher, Arthur Schopenhauer, who had absolute faith in the non-existence of God. Schopenhauer, who took the pessimistic view that human beings have little chance of achieving happiness, believed the aim of life was to renounce the desire for it (Marino, 2004, p.107). And while Nietzsche also propounded the non-existence of God, unlike Schopenhauer, he was “devoted to finding a way of affirming life even though it was without a master plan and planner” (Marino, 2004, p.107).

Nietzsche was the prophet who attacked Christianity and announced ‘the death of God’, and in so doing inspired many of the later existentialists to come to terms with a life lived without a god. For Nietzsche, “it is not only Christ and Christianity that must be rejected in the name of life; it is the very notion of God that must be done away with if man is to be fully man and to assume full responsibility for himself. God must die if man is to live” (Charlesworth, 1975, p.16). Following in Nietzsche’s line of thought, Sartre summarised thus: “if God exists, then man cannot be free; and if man is free then God cannot exist” (Charlesworth, 1975, p.17). Camus questioned the logic that leads to the affirmation of God, asserting that “god is maintained only through the negation of human reason”⁷² (Camus, 2004a, p.470). Sartre and Camus both agreed that in order for people to take themselves and their freedom seriously, they must first reject God; that, in effect, “religion alienates a man from his true self and provides him with a way of evading the full consequences of his freedom” (Charlesworth, 1975, p.17). Kierkegaard also saw religion itself in this way, in that the Church and institutionalised Christianity served to distance a person from their direct relationship with God and the full consequences of such a relationship.

Nietzsche venerated the ‘life-affirming’ spirit of the Greek god Dionysus, seeing it as diametrically opposed to the ‘life-negating’ spirit of Christ. He asserted that the

⁷² It was for this reason that Camus called the existential attitude “philosophical suicide” (Camus, 2004a, p.470).

prospect of living without a God would be terrifying for most because very few have the courage to assume the total responsibility for themselves required to create their own moral values. They prefer to be 'slaves'; to follow the 'herd', he would say. Nietzsche termed the special few who are prepared to live in a life-affirming way – "willing to bear the burden of [their] freedom in Dionysian joy and affirmation" (Charlesworth, 1975, p.17) – the '*Übermenschen*'⁷³, an ideal that was later adopted by Sartre, Camus and Heidegger.

Nietzsche's influence can be seen in Camus' *The Rebel* - one "must learn to live and to die, and in order to be a man, to refuse to be a god" (Camus, 1962, p.269) – and likewise, his influence is clear in Sartre's famous essay entitled, *Existentialism is a Humanism*. In the essay, Sartre commences his line of argument by postulating that if a God does exist, it is plausible to think that the essence of everything is prior to existence – that even "each individual man is the realization of a certain conception which dwells in the divine understanding" (Sartre, 1975, p.348). However, believing it obvious that God does not exist, he was able to claim that

there is at least one being whose existence comes before its essence, a being which exists before it can be defined by any conception of it. That being is man or, as Heidegger has it, the human reality. What do we mean by saying that existence precedes essence? We mean that man first of all exists, encounters himself, surges up in the world—and defines himself afterwards. [...] [H]e will be what he makes of himself. Thus, there is no human nature, because there is no God to have a conception of it. Man simply is. Not that he is simply what he conceives himself to be, but he is what he wills, and as he conceives himself after already existing—as he wills to be after that leap towards existence. Man is nothing else but that which he makes of himself (Sartre, 1975, p.349).

In other words, humans have no pre-existing essence and so define themselves.

⁷³ Often translated as 'supermen', the German word Nietzsche uses actually refers to 'people', not just 'men'.

EXISTENTIALISM...IN THIS STUDY

In this chapter, I have presented a justification of my decision to adopt a particular way of looking at the data in this study – one that is steeped in existential philosophy. It has traced my developing understanding of what would be required of the particular methodological stance that I would eventually take up in relation to my research question. Of utmost importance were that my orientation to the participants be an ethical one⁷⁴ and that the philosophical underpinnings of the approach be congruent with the nature of that which I set out to investigate. Working through the problem of deciding which methodology was required, in turn, deepened my insight of what ‘being a carrier’ is/means in our society. I have shown my arrival at a very different way of looking to examine this from where I had been positioned at earlier stages of the decision-making process – i.e., now favouring a qualitative approach over a quantitative one, an interpretive approach over a positivist one, and, finally, an existential approach over a phenomenological one.

In the families in this study, the genetic state that our society labels as being a ‘carrier’ refers to a tiny sequence change in the DNA of a particular recessive allele⁷⁵ at a single genetic locus in the genome. It is a state that is neither tangible nor apparent to the person ‘afflicted’ by it because they do not sense any manifestation of it or suffer from any symptoms. **It has no intrinsic meaning; is without Being⁷⁶; without essence.** Any meanings attached to it are culturally-derived. It is therefore not a phenomenon that is accessible via the pure form of phenomenology. It only acquires meaning as a social construction of reality, which is why being-a-carrier must be conceptualised as a fundamentally human experience. One needs to focus

⁷⁴ Of course, this meant much more than that my project be approved by an ethics committee. It meant considering how I would position myself in relation to the participants and how the approach to be taken to the study and the kind of findings this would produce would serve to position them.

⁷⁵ The word ‘allele’ refers to alternate forms of a gene (i.e., the many different mutations that exist in the CFTR gene are said to be allelic variations of that gene).

⁷⁶ ‘Being’ (always capitalised) is the term Heidegger used to denote the basic condition of our existence. For him, “[t]he fundamental mystery of life is that something exists, rather than nothing [...] Before anything else—any knowledge, event or thing—the world *exists*” (Lemay & Pitts, 1994, p.32-33). It is “that primordial condition or ‘ground’ which allows [beings] to come into existence” (Lemay & Pitts, 1994, p.34). According to Heidegger, “the world we live in, the world of beings, could only be properly understood in light of existence and non-existence, of Being and Nothing” (Lemay & Pitts, 1994, p.39).

upon the way being-a-carrier manifests in existential terms. One must ask what carriership means in terms of existence and to us as individual human existents⁷⁷.

My aim was clear and compelling for me: interpret how the individual's experience is shaped by what it means to be human; look for the existential categories of experience which pervade the hopes and concerns of parents within carrier families. In order to achieve this, I aimed to devise a 'method' based on the tenets of existential philosophy that could be used to understand my data. In Chapter Three, I delineate a conceptual framework that is derived from this philosophy in fulfilment of this goal.

⁷⁷ Here, the word 'existent' is used as a noun and is synonymous with a 'being in existence'.

CHAPTER THREE: DEVELOPING A CONCEPTUAL FRAMEWORK

THE EXISTENTIALIST 'METHOD' FOCUSES ON CONCRETE EXPERIENCE

Existentialism was "more an intellectual mood or atmosphere than a coherent creed or body of doctrine; more an outlook or "mind-set" than a philosophical "party-line"; more a method or approach than a school of thought" (Charlesworth, 1975, p.1).

Despite the differences in the substance or content of their works, the existentialist philosophers share an interest in common themes (some have already been discussed) but, importantly, they also share a common method – that is, a common approach to problems. This began with their radical break from the rationalistic and systematic idea of philosophy that was to be found in Descartes, Kant and Hegel. It was Kierkegaard's view, in particular, that Hegel's mode of philosophical reflection (trying to "understand the whole of reality within an all-embracing system") was a "distraction from the pressing and important issues of concrete existence [...] leading one away into a realm of fantasy" (as described by Wardlaw, interview with Charlesworth, 1975, p.13-14). Simone de Beauvoir elaborated on this point beautifully, juxtaposing Existentialism's focus on the concrete against the inadequacies of the Hegelian system, reinforcing the need for a practical ethics based solely on the former:

As soon as one considers a system abstractly and theoretically, one puts himself, in effect, on the plane of the universal, thus, of the infinite. That is why reading the Hegelian system is so comforting. I remember having experienced a great feeling of calm on reading Hegel in the impersonal framework of the Bibliothèque Nationale in August 1940. But once I got into the street again, into my life, out of the system, beneath a real sky, the system was no longer of any use to me: what it had offered me, under a show of the infinite, was the consolations of death; and I again wanted to live in the midst of living men. I think that,

inversely, existentialism does not offer to the reader the consolations of an abstract evasion: existentialism proposes no evasion. On the contrary, its ethics is experienced in the truth of life and it then appears as the only proposition of salvation which one can address to men. [...] Regardless of the staggering dimensions of the world about us, the density of our ignorance, the risks of catastrophes to come, and our individual weakness within the immense collectivity, the fact remains that we are absolutely free today if we choose to will our existence in its finiteness, a finiteness which is open on the infinite. And in fact, any man who has known real loves, real revolts, real desires, and real will knows quite well that he has no need of any outside guarantee to be sure of his goals; their certitude comes from his own drive. There is a very old saying which goes: "Do what you must, come what may." That amounts to saying in a different way that the result is not external to the good will which fulfills itself in aiming at it. If it came to be that each man did what he must, existence would be saved in each one without there being any need of dreaming of a paradise where all would be reconciled in death. [emphasis added] (de Beauvoir, 2004, p.435-436)

Kierkegaard's own important legacy to the whole Existentialist movement was his overriding concern about the "fundamental, central dynamics of being human and the strains and stresses within human existence" (Wardlaw, interview with Charlesworth, 1975, p.14). For all the existentialist philosophers,

it is the individual human being who is of central importance and it is the "lived experience" of the individual that is the touchstone of all knowledge. This means the rejection of all "systematic" thought – of the abstract and the necessary and the universal – for the sake of the particular and singular and unique experience of the individual (Charlesworth, 1975, p.9).

Sartre stated that the subject matter of philosophy is solely man-in-the-world or men-in-the-world and the problems that have to be studied can be neatly summarised thus: "man, the world-as-encompassing-man, the relationship of man with the world and of men with one another" (interview with Pierre Vicary cited in Charlesworth, 1975, p.25).

Existentialism emerged as a unique philosophical approach because of this focus upon the individual human existent as the whole object of philosophy; the 'individual human existent' eventually evolving through Heidegger as '*Dasein*'⁷⁸, through Sartre as '*être-pour-soi*' (Being-for-itself), and through Merleau-Ponty as 'man-in-the-world' (Charlesworth, 1975, p.24). According to Wardlaw, the main feature of the 'Kierkegaardian method' (later employed by other existentialists, such as Sartre, Camus and Heidegger) is

its insistence that the important questions facing man are questions that are to be discovered in the concrete details of particular human existence – the human existence of the individual, and the kind of struggles he has [in finding] his standing in the world and [in discovering] a way forward for himself (interview with Charlesworth, 1975, p.13).

As Gabriel Marcel once said, "We do not study problems of philosophy, we *are* those problems" (Charlesworth, 1975, p.9).

The existentialist philosophers called for a return to the unpredictable and unsystematic world of concrete experience. Charlesworth explains that, indeed, for them, concrete experience – or 'lived experience' (*le vécu*⁷⁹) – is the only valid criterion of truth – one must always ask: "What does this mean to me, this individual human existent?" (1975, p.9). And, he says, it is for this reason that existentialists prefer to use a method of philosophising that is "descriptive, or revelatory", continuing that "[t]he philosopher is not so much concerned to explain or to systematize as to evoke, to show, to reveal" (1975, p.9). This is confirmed by a quote from Heidegger: "I cannot *prove* anything in philosophy; but I think I can *show* some things" (n.d. cited in Charlesworth, 1975, p.9), a summary of his assessment that philosophy "is not a matter of explaining or proving or justifying; it

⁷⁸ A Heideggerian term which "denotes 'the manner of Being' possessed by creatures like ourselves" (i.e., self-conscious creatures) (Cooper, 1990, p.67). Translated literally, it means 'being-there'. In order to "understand our lives, our 'average-everydayness'", Heidegger said we must start by conceiving of ourselves, first and foremost, as existing, as "there" (Lemay & Pitts, 1994, p.43).

⁷⁹ Charlesworth attributes the concept *le vécu* to Sartre, however one biography on the life of Simone de Beauvoir indicates that in fact it was an acquaintance of hers, Georges Politzer, who denoted 'that which has been lived' as *le vécu* and that it was to become a major concern for de Beauvoir who puzzled with it "from her earliest works onward" (Francis & Gontier, 1987, p.69).

is rather a matter of evocation, of bringing to light, of making explicit what was implicit, to remind us of what is before our eyes"⁸⁰ (Charlesworth, 1975, p.23). Specifically, for advocates of Existentialism, it is "that elusive and primordial realm of experience that underlies the whole of our existence in the world" which must be rediscovered and made explicit (Charlesworth, 1975, p.23).

It was to this realm of primordial experience that Merleau-Ponty gave the term '*l'être sauvage*' (literally 'wild being' when translated into English),

a primitive level of experience that is prior to all reflection, interpretation and construction, and prior to the distinction that we make upon reflection between the conscious subject and the object of consciousness, the subject being set over against the object (Charlesworth, 1975, p.23).

He set the philosopher the task of evoking and describing this 'pre-reflexive' level of experience. Marcel articulated how this could best be done by using the analogy of a music critic trying to describe a performance of a Beethoven quartet:

Obviously he cannot adequately describe in words what he has heard; nevertheless he can through his words evoke in his readers something of the sense of what he heard. In the same way, the philosopher cannot adequately describe the primordial, pre-reflexive levels of experience that lie behind all other experience; he cannot express in categories that which underlies our categories; but he can [...] "allude" to it (Charlesworth, 1975, p.24).

In order to practise this kind of philosophy, Heidegger necessarily sought to cultivate a new way of thinking. He advocated the adoption of "a kind of meditative, almost passive, prayerful stance before 'being' – so that reality reveals or discloses itself to us in all of its unpredictable variety and richness" (Charlesworth, 1975, p.23). Marcel had a similar outlook. For him,

⁸⁰ This explains why the existentialists see no radical break between philosophy and literature, as "novels, plays, poetry, biography and autobiography, can be just as 'revealing' as formal philosophical analyses" (Charlesworth, 1975, p.9).

reality is not a “problem” to be solved or explained once and for all; rather it is a “mystery” which has progressively and endlessly to be explored and penetrated. The basic philosophical attitude [should be] one of “admiration” or wonder before the mystery of existence (Charlesworth, 1975, p.24).

However, while it has been said (and often celebrated) that the ‘attitudes’ of those like Heidegger and Marcel lead to philosophy that is “of its very nature piecemeal and unsystematic” (Charlesworth, 1975, p.23),

The Existentialist is not an irrationalist in the sense of supporting his claims by appeal to mystical insight, ‘gut’ feeling, or other non-rational founts of knowledge. **He argues, typically, by close description of everyday life, by drawing out people’s own implicit understanding of themselves, and by exposing the incoherence of rival claims.** He proceeds, that is, as a philosopher, not a seer. [emphasis added] (Cooper, 1990, p.14)

This is why many of the existentialists were also novelists. Their arguments are amenable to the form of the novel because they make sense to us in our everyday lives; in our understanding of ourselves and others. We recognise the plausibility of their arguments in the stories they have written. **It is my aim that the plausibility of my argument will resound through the pages of this thesis. In the interviews for this study, I attempt to draw out participants’ “own implicit understanding of themselves”. In analysing the data, I examine any “incoherence” in or between their stories and look to provide an interpretation of how knowledge about genetic information is playing out in their “everyday” lives.**

VALUING THE CONCRETE: A WOMEN’S TRADITION

Despite the fact that almost all existential philosophers were men⁸¹, and all of an era where ‘man’ was used unflinchingly as the generic pronoun, the value they place on the concrete speaks to a feminist ideal. While my focus on the analysis of concrete experience has been grounded in an examination of existential philosophy,

⁸¹ Simone de Beauvoir is our existential heroine!

I also embrace the wisdom of feminist approaches to the conceptualisation of knowledge that further substantiate the credibility of concrete experience.

Patricia Hill Collins is a social scientist who writes on Black feminist thought. In her explication of concrete experience as a criterion for credibility in knowledge claims made by Black women, Collins (1997, p.202) makes reference to a quote by Ruth Shays (n.d.) who, she says, “uses her concrete experiences to challenge the idea that formal education is the only route to knowledge”:

I am the kind of person who doesn't have a lot of education, but both my mother and my father had good common sense. Now, I think that's all you need. I might not know how to use thirty-four words where three would do, but that does not mean that I don't know what I'm talking about....I know what I'm talking about because I'm talking about myself. I'm talking about what I have lived. (cited in Collins, 1997, p.202)

Collins suggests that “[i]mplicit in Ms Shay's self-assessment is a critique of the type of knowledge that obscures the truth, the ‘thirty-four words’ that cover up a truth that can be expressed in three” (1997, p.202). And, she could be seen to be asserting herself as the best historian of her own lived experience – the ‘expert witness’, so to speak.

Collins tells us that **to value the concrete is to value a women's tradition**. She comments that

[s]ome feminist theorists suggest that women are socialized in complex relational nexuses where contextual rules versus abstract principles govern behaviour. This socialization process is thought to stimulate characteristic ways of knowing. [...] Through their child-rearing and nurturing activities, women [...] use the concrete experiences of their daily lives to assess more abstract knowledge claims (1997, p.202).

However, she goes on to cite a study by Luttrell (1989), commenting that while

valuing the concrete may be more representative of women than men, social class differences among women may generate differential expression of this women's value. [Luttrell's] study of

working-class women's ways of knowing found that both white and African-American women rely on common sense and intuition. These forms of knowledge allow for subjectivity between the knower and the known, rest in the women themselves (not in higher authorities), and are experienced directly in the world (not through abstractions) (Collins, 1997, p.202).

So then, perhaps it is no coincidence that a researcher driven to inform the debate about genetic testing by examining the concrete experience of individuals, rather than drawing on abstract ethical principles, should be a woman from a working class background herself!

THE EXISTENTIAL CONCEPTS

*Though they may disagree about the details, the existentialists are linked by their commitment to the common themes of **freedom, choice, authenticity, alienation, and rebellion**⁸². [emphases added] (Marino, 2004, p.xiv)*

With the existential philosophers calling for us to describe things in terms of our experience, all kinds of new 'categories' emerged that allow us to describe, interpret and understand humanity and its idiosyncrasies in a different, and wonder-ful, way. The individual existentialists each tended to focus more on some concepts or 'categories of experience' than others. For Camus, it was the notion of the **Absurd** and its counterpart, **Revolt**; for Heidegger, it was **Angst**, as well as **Care** and the **Technological Attitude; Nausea, Fear** and **Anguish, Bad Faith** and **The Look** were all explored and articulated by Sartre; Simone de Beauvoir condemned us to **Ambiguity**; while Kierkegaard seemed to have a morose obsession with **Despair**. Across the board, existentialists would commonly acknowledge **Freedom** as the supreme value of existentialist thought, while **Authenticity** is seen as its primary virtue (Flynn, 2006, p.79). The experiences these concepts describe are not

⁸² In Camus' writings on this theme, the word has generally been translated into English as 'revolt'.

cognitive – rather, they are a matter of feeling or emotional consciousness (Flynn, 2006, p.23).

Such is the interwoven and overlapping nature of these concepts that in a sense they rely on each other for definition. While this has the potential to lead to seemingly circular definitions, it does reflect the nature of human experience. While Charlesworth refers to the concepts as ‘categories’ of experience⁸³ (1975, p.9), for those who pay regard to semantics this may imply a categorical, reductionist view of experience (by classifying and labelling feelings and emotions, boundaries are formed as they become cleanly defined, setting up an either/or discourse in which categories become exclusive). The reality is that human experience cannot be delineated in this way. And since it is not my belief that the existentialists took this view of experience, for the sake of clarity they will be referred to as ‘existential concepts’ here⁸⁴.

The existential concepts listed above, including the way *Freedom* relates to **Responsibility** and **Choice**, are to be used collectively as an interpretive lens through which to analyse the data from the interviews in this study. The rest of this chapter is dedicated to the delineation of an analytical framework that is based on these concepts, as I have explicated them from my reading of the existentialist literature. For ease of reference, a summary of each concept also appears in the Glossary of Terms at the end of this thesis.

Despair

When, therefore, the existentialist proclaims that the messages of salvation and consolation sanctioned by tradition are no less vain than the hope of fulfilment through worldly pursuits, the ordinary man is doubly offended. Not only his first, but his

⁸³ Cooper referred to them as ‘topics’ (1990, p.127), while Marino differed still in using ‘terms’ (2004, p.xiv) – as can be seen in the heading quote of this section.

⁸⁴ As was stated in the section, ‘A Word on Referencing and Stylistic Matters’, *italics* have been used to denote reference to an existential concept. The word is capitalised where the concept is being referred to in its noun form (i.e., *Nausea*); otherwise it is not capitalised (i.e., *nauseous*).

second, line of defense has been breached. He is in despair. With a logic which is wholly indefensible, though understandable enough, he cries: I am in despair, you have reduced me to despair, therefore you are in despair. But no! the existentialist answers. You were in despair in the first place. It is for that reason you have heard and understood me when I stripped you of your illusions. All that I have done is to make you fully conscious of your despair, and now if you will listen further I will help you master your despair. (Olson, 1961, p.3)

While Kierkegaard describes *Despair* as “a sickness of the spirit, of the self” (2004b, p.41), the concept as it is used by the existentialists does require some clarification. In *Either/Or*, Kierkegaard explains what this *Despair* is by highlighting the fragility of our sorrow and our joy:

He who says that sorrow is the meaning of life has joy outside him in the same way that he who would be joyful has sorrow outside him. Joy may take him by surprise in exactly the same way that sorrow may take the other by surprise. **His life view thus hinges upon a condition which is not in his power**, for it is really just as little in a man’s power to give up being joyful as to give up being sorrowful. **But every life view which hinges upon a condition outside itself is despair.** And so, wanting to sorrow is despair in exactly the same sense as wanting to be joyful, since it always is despair to have one’s life dependent upon that which may pass away. [emphases added] (1959, p.240)

We see that this reference to *Despair* is not to a mood of hopeless gloom, but rather to the position of someone whose life, while contented perhaps, “hinges upon a condition outside itself”.

Sartre adapts this into an atheistic concept in *Existentialism is a Humanism*, where he says that *Despair* brings the recognition that we should

limit ourselves to a reliance upon that which is within our wills [...] Beyond the point at which the possibilities under consideration cease to affect my action, I ought to disinterest myself. For there

is no God and no prevenient design, which can adapt the world and all its possibilities to my will (Sartre, 1975, p.357).

So, in *Despair* I come to the realisation that I am largely ineffectual in terms of the influence I have on much of the greater world about me. As Sartre says, in order to effect change, "whatever may be in my power to make it so, I shall do; beyond that, I can count upon nothing" (Sartre, 1975, p.358).

A reading of the sections 'Despair Is the Sickness unto Death', 'The Universality of This Sickness (Despair)' and 'The Forms of This Sickness (Despair)' from his treatise, *The Sickness unto Death* (Kierkegaard, 2004b, p.41-105), reveals that for Kierkegaard there is one mode of human existence only and that is *Despair*. In short, he says, if someone does not think they are in *Despair*, they undoubtedly are. If they think they are in *Despair*, they are probably wrong about what they think their *Despair* is - they are likely to be in *Despair* but not in the way that they think. We are lucky to be able to *despair* (its possibility indicates our 'superiority' over the animal) but tortured by our *Despair* (its actuality is not only the worst misfortune and misery - it is also 'ruination'). Men and women *despair* in different ways: women *despair* when they lose their object of devotion for they do not will to be themselves (they wish to lose themselves by focusing their devotion outside of themselves, in their feminine way) while men *despair* in willing to be themselves (it is not masculine to be able to lose themselves in their devotion).

In spite of his 'morose obsession', a conclusion regarding the effect this must have on those drawn to Kierkegaard's philosophy should not be drawn too quickly for there is a surprise to be found here. For all of Kierkegaard's convoluted prosaic ramblings around *Despair*, there are readers who have drawn deep inspiration from his work (in particular, the magnificent distinction he drew between *Despair* and depression⁸⁵). For the professor of philosophy, Gordon Marino, who had been "in

⁸⁵ Kierkegaard's view of depression, that it is "marked by inexplicable sadness and self-loathing", matches that of modern psychology. However, in the present age, the distinct nature of *Despair* has not been preserved. For Kierkegaard, the depressed individual is also in *Despair* if they "[see] quite clearly that this depression ... is of no great significance" and it is precisely that fact, "that it neither

the cold grip of a withering depression”, Kierkegaard helped “to make the turn from thinking of suffering as a fever to regarding it as an action that could be carried out with dignity” (Marino, 2004, p.x).

Self-estrangement (a form of alienation) versus Authenticity

If existentialism is a philosophy to live, it is to be lived by men and women who have not become estranged from themselves (Cooper, 1990, p.36).

According to two other philosophers, Hegel and Marx, there are various modes of alienation – alienation from the world, from others, and from oneself. Alienation is a key consideration in existential thought. The existentialists blame our tendency toward ‘dualistic illusions’ of thought (i.e., subject versus object, mind versus body, and fact versus value) for our sense of alienation from the world (Cooper, 1990, p.35-36).

Heidegger (1996) tackles these dualisms in his philosophical treatise, *Being and Time*, while a constant theme in his later writings on Technology is “our ‘estrangement from Being’”, where people are described as “‘homeless’⁸⁶ and in search of a true ‘dwelling’” (Cooper, 1990, p.35). Existentialists believe that people generally respond to this threat of ‘homelessness’ and “suppress a sense of alienation from the world by becoming ‘absorbed’ in or ‘tranquillized’ by the comforting, ready-made schemes of beliefs and values which prevail in their societies” (Cooper, 1990, p.33). According to Flynn, Heidegger argues that this occurs *en masse* because

we are for the most part immersed⁸⁷ in the average everyday where the inclination is to neglect our openness to Being and to

has nor acquires any great significance”, that it is *Despair* (Kierkegaard n.d. cited in Marino, 2004, p.x).

⁸⁶ Cooper has explained this concept of ‘homelessness’ as being “abandoned in a world devoid of meaning and value, a world whose ‘unimaginable otherness’ is, at best, partly accessible only to the man in the laboratory” (1990, p.36).

⁸⁷ It is this immersion that Heidegger calls our ‘fallenness’ (*die Verfallenheit*) (Flynn, 2006, p.70).

simply 'go with the flow', that is, to live inauthentically as 'they' do (Flynn, 2006, p.70).

But Jaspers urges the individual to struggle against this kind of 'mass-existence'; in his words:

Although my social I is [...] imposed upon me, I can still put up an *inner* resistance to it. [...] Although I am in my social I at each moment, I no longer coincide with it. [...] I am not a result of social configurations, [...] [for] I retain my own original potential. (Jaspers, 1970, p.30)

Failure to resist one's "social I" constitutes loss of self⁸⁸; according to Heidegger, it is in the 'they' that a person is at risk of losing their "ownmost potentiality"⁸⁹ for being-in-the-world" (Heidegger, 1996, p.166). And when *Dasein's* "ownmost potentiality for being-in-the-world is concealed [...], this alienation [...] *closes off* to Da-sein its authenticity and possibility" (Heidegger, 1996, p.166).

The existentialists refer to the kind of existence where an individual's *Existential Freedom* is not exercised as an *inauthentic* one; they have become estranged from themselves. Thus, the concept of *Self-estrangement* is virtually equivalent to that of *Inauthenticity*. *Authenticity*, on the other hand, is for Sartre a kind of 'self-recovery of being'; a recovery of that which has been lost during *Self-estrangement* (Cooper, 1990, p.101). He borrowed the term '*Authenticity*' (*Eigentlich*) from Heidegger but, despite Heidegger's protests that it was not endowed with any moral significance, Sartre insisted that it was (Flynn, 2006, p.65). This is because an *authentic* existence, in which an individual overcomes *Self-estrangement* and, as Kierkegaard would say, "become[s] an individual", "requires a person to disengage himself from the ways of the 'Public', the 'herd' or the 'they'⁹⁰" (Cooper, 1990, p.33) and this

⁸⁸ For a sample of Karl Jaspers' writing on "my being as a social I" and loss of self due to immersion in the 'they' (1970, p.30), see entry for **(The) 'they'** in the Glossary of Terms.

⁸⁹ This word was alternatively translated as 'possibilities' in the English translation of *Sein und Zeit* published by Blackwell in 1962, and perhaps allows a more intuitive understanding of what Heidegger means here. I use the term 'possibilities' throughout the rest of this thesis.

⁹⁰ The 'they' is the usual translation of the Heideggerian term '*Das Man*', an expression using the indefinite pronoun '*man*' (equivalent to the French '*on*') (Cooper, 1990, p.112). While Heidegger favoured this term, Kierkegaard preferred using the 'Public' and Nietzsche the 'herd' to express the same idea (Cooper, 1990, p.14).

requires a brave choice by an above-average individual. Nietzsche spoke of the loneliness of the person who manages to rise above the 'herd', remarking⁹¹:

Today ... when only the herd animal is honoured ... the concept of 'greatness' entails being noble, wanting to be oneself, being capable of being different, standing alone and having to live independently (cited in Flynn, 2006, p.25).

This contrast between the life of the *authentic* individual and the life which is immersed in the anonymous 'they' is a theme at the heart of Existentialism. All existentialists would concur with the notion that a person's existence is an 'issue' for them. And in confronting this, each person "must develop beliefs, values, and interpretations of his situation, which will direct the shape he gives to his life" (Cooper, 1990, p.114). The person may "drift with the prevailing breeze"⁹² (Cooper, 1990, p.114) but in so doing, Heidegger says they live "under 'the dictatorship of the "they"', the 'anonymous others'", and fail to make their life their own (Cooper, 1990, p.111); they live an *inauthentic* existence.

Existentialists see the pull in modern society as being "away from individualism and towards conformity", and so they regard being an existential individual in our 'mass society' as "an achievement rather than a starting point" (Flynn, 2006, p.24). They view the achievement of *Existential Authenticity* as

a task to be undertaken and sustained but perhaps never permanently achieved. [...] [T]he time-bound nature of the human condition requires that existing as an individual is always dynamic and under way, never static and complete. And depending on the circumstances, it may also involve considerable risk (Flynn, 2006, p.24-25).

The risk is present because a withdrawal from the 'they' has the potential to bring a person into conflict with others. However, existentialists hope that their philosophy is one through which it is possible for people to view themselves as "'at home' with their world and each other, but not at the cost of 'losing themselves'" (Cooper, 1990, p.33). Living *authentically*, according to Nietzsche, is the *Urbemensch* who, in

⁹¹ Precise reference not given.

⁹² Cooper likens this behaviour to the 'aesthetic' figures described in Kierkegaard's *Either/Or* (1959).

“realiz[ing] to what degree we are the *creators* of our value feelings—and thus capable of projecting ‘meaning’ into history” (Nietzsche, 1968, s.1011) – is afforded the power that comes from being able to lend ‘sense and value’ to his life and his world. And, “[i]n so doing, he represents in the most vivid way the essentially creative and interpretative character of human existence” (Cooper, 1990, p.112).

Bad Faith and the ‘Predominance of the Other’

[W]e need to describe in some detail those modes of existence which, according to The Existentialist, are ones of inauthenticity or self-estrangement. These modes are made possible by our Being-with and our Being-for others (Cooper, 1990, p.110).

Sartre emphasises a different mode of *Self-estrangement* – the *Predominance of the Other* – and this is a recurrent theme in his fiction. It is also the main theme in what is perhaps the most famous existentialist read, de Beauvoir’s *The Second Sex*. Those who are self-estranged through the *Predominance of the Other* identify too fully with their Being-for-others and Simone de Beauvoir argues that woman is guilty of conceiving of herself as ‘other’ because her perspective of herself is centred too much around the male view of her.

Cooper paraphrases Sartre from *Cahiers pour une Morale*⁹³ and describes *Self-estrangement* through the *Predominance of the Other* thus:

A person gets ‘taken hold of by others’, in the sense of coming to construe himself as he is for others. He ‘conceives his own consciousness on the model of the Other’, as if he were to himself as he is to others and they to him [...] [He] *impersonates* himself, by identifying too completely with the role in which others see him (Cooper, 1990, p.117).

In *Being and Nothingness*, Sartre uses the case of the ‘waiter in the café’ as an example:

⁹³ *Notebooks for an Ethics* is the English translation of this title.

His movement is quick and forward, a little too precise, a little too rapid. He comes toward the patrons with a step a little too quick. He bends forward a little too eagerly; his voice, his eyes express an interest a little too solicitous for the order of the customer. Finally there he returns, trying to imitate in his walk the inflexible stiffness of some kind of automaton while carrying his tray with the recklessness of a tight-rope-walker [...] All his behaviour seems to us a game. [...] He is playing, he is amusing himself. But what is he playing? We need not watch long before we can explain it: he is playing at being a waiter in a café. [...] The game is a kind of marking out and investigation. [...] [T]he waiter in the café plays with his condition in order to realize it (1958, p.59).

In acting out this 'social role'⁹⁴, the waiter "[has 'lost'] what is unique about himself, and in that sense ['lost'] his very self" by

identifying too much, and too easily, with the 'communal character' of his existence. [...] He no longer 'owns' himself since, in one way or another, he has succumbed to a take-over by others [...], he is not authentic (Cooper, 1990, p.109).

Flynn too is articulate in his assessment of Sartre's waiter and why being "a waiter with his entire being, the way a stone is a stone" is living *inauthentically*:

He has become the slave to an image that others' expectations have imposed upon him and which he has appropriated. Bad faith [has entered because he] dismisses any other kind of behaviour as inconceivable (Flynn, 2006, p.73).

According to Cooper,

The Existentialist cannot accept that life's 'communal character', such as the playing of 'social roles', is even approximately the whole story about human existence. [...] What The Existentialist does accept is that people are only too liable to live *as if* the 'communal character of existence' were the whole story ... [and] when they do that their lives are self-estranged (Cooper, 1990, p.110).

⁹⁴ While the example chosen by Sartre happens to focus on an occupational role, of course there are many other social roles, predicated by such things as gender, race, sexuality, birth order, etc., that a person can 'play'.

Sartre describes our Being-for-others as “a fall through absolute emptiness toward objectivity” – toward our own objectification – and, of course, “this fall is an *alienation*” (1958, p.274-275). In self-reflection “[u]pon any one of my conducts it is always possible to converge two looks, mine and that of the Other” (Sartre, 1958, p.57); we can see ourselves through our own eyes or as we are through the eyes of others. And “[b]ecause of this, I may exclude from my self-rumination any help that others may provide, convinced that they are ‘possessed only of a deformed image of me’” or, “[a]lternatively, I might focus exclusively on how I am for others, in the belief perhaps that any solo feats of self-analysis would be a wayward exercise in narcissistic introspection” (Cooper, 1990, p.118).

In *Self-estrangement through the Predominance of the Other*, a person comes to adopt largely the third-person perspective of themselves. Cooper describes what this means for such an individual:

He comes to view himself as a series of events in the world, no different in principle from the series of events that constitute the causal histories of physical objects [...] [T]his person [views] himself as a ‘substance’, and not as an ‘existing individual’ for whom causal, probabilistic modes of explanation are inappropriate (Cooper, 1990, p.121).

According to Sartre, *Self-estrangement through the Predominance of the Other* is a form of *Bad Faith*: “I recognize the Other as the subject through whom my being gets its object-state” but if “I [...] resign myself to *being only that*”, this is “a reaction of [...] bad faith” (Sartre, 1958, p.290). Here, the person identifies “too fully” with their “image for others” instead of with “a more solipsistically formed self-image” (Cooper, 1990, p 118). Sartre believed that “[t]oo many of us [...] take the words of others as the last judgement” (Cooper, 1990, p.121). Clearly, our tendency to this form of *Bad Faith* is inherent in the very fact of our Being-for-others (Cooper, 1990, p.120). Societies are eager for people to accept, and live according to, the label given to them by others because their behaviour then becomes amenable to prediction and, hence, modification – a comfortable state of affairs for the ‘they’.

Flynn defines Sartre's concept of *Bad Faith* as "the spontaneous ordering of one's life to settle on non-persuasive evidence"⁹⁵ (2006, p.72), while Cooper tells us that it is generally connected with self-identity; with a person's conception of who they are and what they are like (1990, p.118). Cooper summarises the other forms that *Bad Faith* can take as "paying no heed at all to how one is for others"⁹⁶, "identifying with one's 'objective' body as something to which things simply happen"⁹⁷, and "identifying too closely with how one *has* been, with one's past, thereby divesting oneself of responsibility for one's future"⁹⁸ (1990, p.117).

A reading of *Being and Nothingness* (Sartre, 1958) reveals that *Bad Faith* is a lie to oneself; it is from myself that I am hiding the truth when I live in *Bad Faith*; it is an act of self-deception. Ordinarily, says Sartre (1958, p.49), the lie is a normal phenomenon of Being-with-others in the world: "[i]t presupposes my existence, the existence of the *Other*, my existence *for* the Other, and the existence of the Other *for* me". A lie exists by its very nature as "*hidden from the Other*; it utilizes for its own profit the ontological duality of myself and myself in the eyes of the Other". In contrast, "the duality of the deceiver and the deceived does not exist [with *Bad*

⁹⁵ Flynn says that Sartre classifies knowledge as 'certain' when its object can be "grasped reflectively in an immediate 'self-evident' intuition" (persuasive evidence), distinguishing this from belief which is only 'probable' because "the object pursued is merely indicated by something else that is evidence for or against its presence" (non-persuasive evidence) (2006, p.71).

⁹⁶ Despite Sartre urging us to reject the *Predominance of the Other*, we would be foolish to completely ignore how others regard and assess us. It is a thin line "between paying proper heed to what others say to us and about us, and surrendering before the view they take of us" (Cooper, 1990, p.121).

⁹⁷ In this pattern of *Bad Faith*, a person conceives of their body as Being-in-itself (translation of Sartre's term, *être-en-soi*, for non-conscious Being), and is thus "relieved of a sense of responsibility for [their] life" (Cooper, 1990, p.121). The person, while sensing the presence of their own body, does not consider themselves as **being** their own body – they contemplate it "as though from above as a passive object to which events can *happen* but which can neither provoke them nor avoid them because all its possibilities are outside of it" (Sartre, 1958, p.56). Sartre uses the example of the woman on a first date with a particular man: when he first takes her hand, she leaves it there but "she *does not notice* that she is leaving it [...] because it happens by chance that she is at this moment all intellect. [...] [S]he speaks of Life, of her life, she shows herself in her essential aspect—a personality, a consciousness. And during this time the divorce of the body from the soul is accomplished; the hand rests inert between the warm hands of her companion—neither consenting nor resisting—a thing" (Sartre, 1958, p.55-56).

⁹⁸ The person living in this kind of *Bad Faith* is transfixed by the 'character' which other people have assigned to them based on a summary of the tendencies of their life as so far lived. According to Sartre, this is "tantamount to handing oneself over to the verdict of others" (Cooper, 1990, p.120), putting constraints on their future possibilities rather than remaining open to the transformative effect that the possibility of future choices and future actions could bring. Thus, they have lost their *Freedom*.

Faith. *Bad faith* [...] implies in essence the unity of a *single* consciousness". It should be noted here that while *Bad Faith* may at first glance appear to be synonymous with the notion of 'living in denial' generally, it is in fact a rather specific form of denial – denial of responsibility for one's situation (Flynn, 2006, p.70).

The person living in *Bad Faith* must know the truth in order to conceal it more carefully from themselves (Sartre, 1958, p.49). And yet, in doing this,

there is no question of a reflective, voluntary decision, but of a spontaneous determination of our being. One *puts oneself* in bad faith as one goes to sleep and one is in bad faith as one dreams (Sartre, 1958, p.68).

According to Cooper,

the more effective one's strategies of bad faith, the more complete one's unreflecting 'absorption' in the 'they', the less capable is one of recognizing the state of inauthenticity. Like Kierkegaardian 'despair', existentialist self-estrangement is all the more entrenched to the degree that it is unaware of itself as such (1990, p.123).

And it follows that

[o]nce this mode of being has been realized, it is as difficult to get out of it as to wake oneself up; bad faith is a type of being in the world, like waking or dreaming (Sartre, 1958, p.68).

However, "because of the unblinking eye of pre-reflective consciousness", a person is aware that they have ordered their life according to "non-persuasive evidence" and so "remains responsible for remaining in bad faith" (Flynn, 2006, p.72).

The Look

'The Look' is at work whenever I am made aware of myself as an object for the attention of others: creatures who can 'transfix' me in the way I 'transfix' objects about me (Cooper, 1990, p.105).

A human being can only experience emotions such as shame, modesty, pride, shyness, loss of face, and dignity because they have a sense of being “an individual, a distinct presence in the world” – this arising through their “‘concrete’ relations with others” (Cooper, 1990, p.104-105). In the section on ‘Being-for-Others’ in his book, *Being and Nothingness*, Sartre (1958) introduced the notion of *The Look* (*le regard*) as the root source of quintessentially human experiences such as these. In his words, “the look has set us on the track of our being-for-others and has revealed to us the indubitable existence of this Other for whom we are” (1958, p.282). He uses as an example our experience of shame: in reflecting upon my experience of shame, I realise that “I am ashamed of myself before the Other” (1958, p.289).

Sartre created a vignette to illustrate *The Look* with his typical dramatic flair:

Let us imagine that moved by jealousy, curiosity, or vice I have just glued my ear to the door and looked through a keyhole. I am alone [...] [b]ut all of a sudden I hear footsteps in the hall. Someone is looking at me! What does this mean? It means that I am suddenly affected in my being and that essential modifications appear in my structure – modifications which I can apprehend and fix conceptually by means of the reflective *cogito* (1958, p.259-260).

Cooper borrows Sartre’s own words (1958) to explain that

[t]he crucial modification [here] is that ‘I now exist as myself’, for I have been made into ‘an object for the Other’. ‘The Look’ at once reifies and individuates me. ‘[B]ehold now I *am* somebody’. (1990, p.105)

Whereas moments before his “mode of being was governed by unreflective consciousness”, “an essential change occurs in [his] mode of awareness” such that he experiences himself as an object for the other (van Manen, 1990, p.25). Thus, in the realisation that he is being looked at, Sartre’s man-at-the-keyhole “experiences his body ‘objectified’ by another consciousness” (Flynn, 2006, p.21) and he is mortified by this:

Becoming apprised of ‘the Look’ is, for Sartre, only the beginning of one’s sense of distinctive selfhood. [...] [B]eing subject to ‘the

Look' is a disturbing experience because, being a free, spontaneous For-itself, I cannot be the mere object – the squatting voyeur at the keyhole – to which 'the Look' threatens to reduce me. Hence, by way of self-defence, I engage in 'a refusal of the Other'; and in the ensuing battle, during which I reaffirm my subjectivity against the other, 'I ... obtain an explicit self-consciousness [through] a negation of the Other' (Cooper, 1990, p.105-106)⁹⁹.

In other words, our indignant reaction to the realisation that we have become depicted as "object" is to re-assert our subjectivity.

According to Sartre, "my 'primordial' experience of others is [...] 'Being-for' them as an object of their attention" (Cooper, 1990, p.106). By this, he means that we do not become aware of the presence of another person looking in our direction and then become conscious of ourselves in their eyes upon reflection, rather it is the other way around – **we are first aware that we are subject to *The Look* and then reflect upon who it is looking at us.** The following passage from *Being and Nothingness* elaborates this point:

[F]ar from perceiving the look *on* the objects which manifest it¹⁰⁰, my apprehension of a look turned toward me appears on the ground of the destruction of the eyes which "look at me". If I apprehend the look, I cease to perceive the eyes [...] [W]e can not (*sic*) perceive the world and at the same time apprehend a look fastened upon us; it must be either one or the other. This is because to perceive is to *look at*, and to apprehend a look is [...] to be conscious of *being looked at*. [...] What I apprehend immediately when I hear the branches crackling behind me is not that *there is someone there*; it is that I am vulnerable, that I have a body which can be hurt, that I occupy a place and that I can not (*sic*) in any case escape from the space in which I am without defense – in short, that I *am seen*. Thus the look is first an

⁹⁹ Here, Cooper cites Sartre (1957).

¹⁰⁰ Sartre had earlier clarified that "[o]f course what *most often* manifests a look is the convergence of two ocular globes in my direction [...] [but it can just as readily be] a rustling of branches, or the sound of a footstep followed by silence, or the slight opening of a shutter, or a light movement of a curtain" (Sartre, 1958, p.257) – "it is never eyes which look at us; it is the Other-as-subject" (Sartre, 1958, p.277).

intermediary which refers from me to myself (Sartre, 1958, p.258-259).

So, from an existential perspective, what is important about *The Look* is not so much my awareness of the person looking but my consciousness of feeling vulnerable and exposed.

Sartre is convincing in his argument that it is in the eyes of the Other we come to know ourselves, continuing to use shame as his exemplar:

It is certain that my shame is not reflective, for the presence of another in my consciousness, even as a catalyst, is incompatible with the reflective attitude; in the field of my reflection I can never meet with anything but the consciousness which is mine. But the Other is the indispensable mediator between myself and me. I am ashamed of myself *as I appear* to the Other.

By the mere appearance of the Other, I am put in the position of passing judgment on myself as on an object, for it is as an object that I appear to the Other. [...] Shame is by nature *recognition*. I recognize that I *am* as the Other sees me. [...] But this new being which appears *for* the other does not reside *in* the Other; I am responsible for it [...] Thus shame is shame *of oneself before the Other*; these two structures are inseparable. But at the same time I need the Other in order to realize fully all the structures of my being (Sartre, 1958, p.221-222).

So, what Sartre is saying here is that it is through my relation with the Other that I realise my own shame. Without the Other, this shame would not have been awakened within me. *The Look* is a mechanism for bringing the, at times, deep-seated contents of my consciousness to the surface such that they are recognised and experienced by me as a certain emotion. In *The Look*, I become aware of myself through my sudden recognition of how I appear to the Other.

Angst¹⁰¹

Existentialism is said to have given voice to an *Angst* (and *Despair*) which was “peculiarly symptomatic of the twentieth-century condition” (Cooper, 1990, p.13). The *Angst* of which existentialists speak does not refer to “that fear before a dangerous, uncertain world” which, Cooper says, Virginia Woolf (1985) alludes to in her *Diaries*:

The war ... has taken away the outer wall of security. No echo comes back. I have no surroundings. Those familiar circumvolutions ... which have for so many years given back an echo ... are all wide and wild as the desert now. (cited in Cooper, 1990, p.13-14)

Instead, Cooper says:

Existential *Angst* is, rather, a sense of freedom, of a capacity to strike out on one’s own in the formation of a scheme of beliefs and values. If *Angst* has special significance in modern times, this is not because life has become too ‘dishevelled’ or ‘wide and wild’, but because it has become too *comfortable*. Beliefs and values are too easily and readily received from [...] the ‘they’ (1990, p.14).

Our experience of *Angst* springs from our pre-philosophical “implicit awareness of our freedom as the sheer possibility of possibility” (Flynn, 2006, p.70).

According to Cooper, it is useful to distinguish two stages of *Angst* in order to understand why a person’s relationship to *Angst* can be, in one instance, troubling and, in another, a positive experience. The first way that *Angst* may be experienced is as a “disturbing and ‘uncanny’ mood which summons a person to reflect on his individual existence and its ‘possibilities’” (Cooper, 1990, p.128). Cooper says “[t]his

¹⁰¹ There are many words used in the existentialist literature to refer to this phenomenon. Cooper (1990, p.127) favours staying with the German word, ‘*Angst*’, over the English words offered in translation, such as ‘anxiety’ and ‘dread’, and over expressions used by other existentialists, such as ‘anguish’ by Sartre and ‘metaphysical fear’ by Jaspers. For simplicity, I do the same in this section. In the next section, where this concept is explicitly contrasted with *Sartrean Fear*, I invoke the term, *Anguish*, because I rely heavily on Sartre’s differentiation of the two. I believe his explication of the two concepts as they relate to free will and determinism has significant parallels to our paradoxical relationship with nature and Technology, which is why I continue to refer to these concepts as *Sartrean Fear and Anguish* when discussing these issues in later chapters of this thesis.

Angst is not something to be 'treated'" (1990, p.14) but that we generally endeavour to avoid such *Angst* "by 'fleeing' into bad faith and the comforting embrace of the 'they'" (1990, p.128). He says that, instead of doing this, "we need to be called to [*Angst*], and away from a state of 'tranquillization' induced through bad faith" (1990, p.14). What this requires is that we "[face] up" to our *Angst*, and accept the truths about our existence which it intimates (1990, p.128). This will imply different consequences (different ways of living out their *Angst*) for different people depending on how they individually extrapolate these 'truths'.

Vertigo is an excellent metaphor for *Angst*, and has been used by existentialists to conjure up that recognisable feeling of 'groundlessness' that is common to both experiences. Cooper has summarised some of the analogies that have been made between the two:

Kierkegaard refers to [*Angst*] as the 'dizziness of freedom'¹⁰², while Jaspers describes metaphysical fear as 'the vertigo and trepidation of freedom facing a choice'¹⁰³. **Vertigo requires the presence of an abyss, the absence of a supporting ground. *Angst* likewise is the experience of groundlessness, the absence of anything holding one in place and anchoring one's actions.** At a micro-level, it is the ex-gambler's realization that nothing 'secures' his vow to abstain. At a macro-level, it is Virginia Woolf's 'tragic' sense of life as 'a strip of pavement over an abyss'.¹⁰⁴ [emphasis added] (1990, p.130)

So, through the use of vertigo as a metaphor, we better understand how *Angst* is, then, the appreciation that none of the exigencies, values and commitments with which we find ourselves embroiled in everyday life furnishes sufficient grounding for the attitudes, interpretations and behaviour we adopt. [...] At its deepest and most dramatic, *Angst* discloses 'a being which is compelled to decide the meaning of being'¹⁰⁵ (Cooper, 1990, p.130).

¹⁰² Cooper cites Kierkegaard (1980).

¹⁰³ Cooper cites Jaspers (1970).

¹⁰⁴ Cooper cites Woolf (1985).

¹⁰⁵ Here, Cooper cites Sartre (1957).

So, for Sartre, it is in the *Freedom* disclosed by this *Angst* that a person realises they need only be moved by those “appeals of the world” which they permit themselves to be moved by (Cooper, 1990, p.130). And this is why Heidegger says that *Angst* individualises *Dasein* (1996, p.176) – because each person’s understanding of themselves and their world can no longer be unthinkingly restricted to the familiar interpretations offered by the ‘they’ (Cooper, 1990, p.131).

Sartrean Fear and Anguish (Angst)¹⁰⁶

Heidegger described *Angst* and ordinary fear as “kindred phenomena” (1996, p.173) but it was Kierkegaard who inspired the distinction that many existentialists agree may be drawn between the two. Sartre was one of those in agreement, differentiating them thus: “fear is fear of beings in the world whereas anguish is anguish before myself” (1958, p.29). Flynn has elaborated on this:

Whereas fear has a definite object, [...] [anguish] is the awareness that any choice is within our power to make, even if its success may elude us. [...] The possibilities to which [anguish] refers, even the possibility with no specific object – the sheer awareness of freedom – denotes the consciousness of *my* situated freedom and possibility (2006, p.70-71).

We *fear* a certain thing, whereas Heidegger says that “*Angst* ‘does not know’ what it is about which it is anxious” (1996, p.174). However, even though *Angst* is distinct from *Fear* because it is not directed at particular objects and events, *Angst* is still always anxiety about something – it is never undirected. Like any mental state, *Angst* always “has an object in the sense of being *towards* or *about*” but this object is not one “in the sense of a spatio-temporally located particular” (Cooper, 1990, p.129). For example, Heidegger states it is “*the world as such*” or “*being-in-the-world itself*” (1996, p.175). The various existentialists have speculated upon other possible objects of this *Angst* in addition to those already mentioned (i.e., “myself” as put forward by Sartre and “the world” and “being-in-the-world” according to Heidegger) and those proposed include: “my freedom, nothingness, emptiness, [...] finitude, and death” (Cooper, 1990, p.129).

¹⁰⁶ The special differentiated use of the terms *Angst* and *Sartrean Anguish* in this thesis was explained in a footnote earlier in this chapter (see section: *Angst*).

According to Cooper, it was Heidegger more than the other existentialist writers who recognised the strategic importance of *Angst*. He quotes from Heidegger¹⁰⁷ to illustrate this:

If *Dasein* is not doomed to 'lose' itself in the 'they', there must be 'a way of disclosure in which *Dasein* brings itself before itself' [and] [t]his way is *Angst* (Cooper, 1990, p.130).

In *Angst*, Heidegger says that

the things at hand in the surrounding world sink away, and so do innerworldly beings in general. [...] It throws Da-sein back upon that for which it is anxious, its authentic potentiality-for-being-in-the-world. (1996, p.175-176)

Angst, he explains, "fetches Da-sein back out of its entangled absorption in the 'world'" and "[e]veryday familiarity collapses" (1996, p.176). Far from *fearing* particular objects and events, they are now "completely without importance"; irrelevant; "[t]he world has the character of complete insignificance" (Heidegger, 1996, p.174).

Charlesworth likened Sartre's consideration of our experience of *Fear* and our experience of *Anguish* to a discussion of the classical problem of free will and determinism (1975, p.9). In *Being and Nothingness*, Sartre says that

[v]ertigo is anguish to the extent that I am afraid not of falling over the precipice, but of throwing myself over. A situation provokes fear if there is a possibility of my life being changed from without; my being provokes anguish to the extent that I distrust myself and my own reactions in that situation. [...] [T]he recruit who reports for active duty at the beginning of the war can in some instances be afraid of death, but more often he is "afraid of being afraid;" that is, he is filled with anguish before himself. [...] [F]ear and anguish are exclusive of one another since fear is unreflective apprehension of the transcendent and anguish is reflective apprehension of the self; the one is born in the destruction of the other (1958, p.29-30).

¹⁰⁷ Martin Heidegger, *Being and Time*, Blackwell, 1962, p.182H.

Sartre uses vertigo as a metaphor not only for delineating *Anguish* but also for differentiating it from *Fear*:

Vertigo announces itself through fear; I am on a narrow path—without a guard-rail—which goes along a precipice. The precipice presents itself to me as *to be avoided*; it represents a danger of death (1958, p.30).

At this point Sartre (1958, p.30) talks about the *Fear* that overcomes this person as they become aware that a number of adverse events, “originating in universal determinism”, could befall them and “transform that threat of death into reality” (i.e., they could slip and fall over the edge or the path could crumble beneath them). He says the person now sees themselves “as a thing”, “an object in the world” and therefore “passive in relation to these possibilities” (1958, p.30). The person reacts to this *Fear* by reflecting on what they need to do to stay safe and to “keep the threats of the world at a distance from [them]” (1958, p.30). These potential future conducts are their possibilities – whether the person engages in them or not will be a matter for their own free will. And in knowing this – that “any conduct on [their] part is only *possible*” (Sartre, 1958, p.31) – the person now experiences *Anguish*. This *Anguish* is exacerbated in the realisation that it is a matter of their free will alone that prevents them enacting another possibility before them – to throw themselves over the precipice. Let us return now to the thoughts of Sartre’s dare-devil:

It is through my horror that I am carried toward the future [...] *Anguish* is precisely my consciousness of being my own future, in the mode of not-being. If *nothing* compels me to save my life, *nothing* prevents me from precipitating myself into the abyss. The decisive conduct will emanate from a self which I am not yet. Thus the self which I am depends on the self which I am not yet to the exact extent that the self which I am not yet does not depend on the self which I am. Vertigo appears as the apprehension of this dependence (1958, p.32).

So, in summary, *Fear* is “the experience we have when we contemplate being caught in the deterministic process of nature” and *Anguish* is “the experience we

have when we realize our total responsibility for ourselves" (Charlesworth, 1975, p.9).

Nausea

Sartre characterised the concept of *Nausea* as "our experience of the contingency of existence" – the knowledge that we are and that we need not be (Flynn, 2006, p.7). Unique to human beings – because "[o]nly humans ponder why they exist at all" (Flynn, 2006, p.58) – Sartre depicted *Nausea* as a 'phenomenon of being' (Flynn, 2006, p.7). This, he illustrated in his philosophical novel of the same name via a soliloquy by the main character:

The Nausea hasn't left me and I don't believe it will leave me for quite a while; but I am no longer putting up with it, it is no longer an illness or a passing fit: **it is me.** [emphasis added] (Sartre, 1963, p.182)

In his novel entitled *Nausea*, Sartre exposes the thoughts of his protagonist as he contemplates the root of a chestnut tree, which is "plunged into the ground" just underneath the bench on which he is sitting in a municipal park:

I no longer remembered that it was a root. Words had disappeared, and with them the meaning of things, the methods of using them, the feeble landmarks which men have traced on their surface. [...] And then I had this revelation.

It took my breath away. Never, until these last few days, had I suspected what it meant to 'exist'. [...] Even when I looked at things, I was miles from thinking that they existed: they looked like stage scenery to me. I picked them up in my hands, they served me as tools [...] If anybody had asked me what existence was, I should have replied in good faith that it was nothing, just an empty form which added itself to external things, without changing anything in their nature. And then, all of a sudden, there it was, as clear as day: existence had suddenly unveiled itself. It had lost its harmless appearance as an abstract category: it was the very stuff of things, that root was steeped in existence. [...]

[...] [All] objects ... how can I explain? They embarrassed me; I would have liked them to exist less strongly, in a drier, more abstract way, with more reserve. [...] We were a heap of existents inconvenienced, embarrassed by ourselves, we hadn't the slightest reason for being there, any of us, each existent, embarrassed, vaguely ill at ease, felt superfluous in relation to the others. *Superfluous*: that was the only connexion I could establish between those trees, those gates, those pebbles. [...] *Superfluous*, the chestnut tree, over there, opposite me, a little to the left. *Superfluous*, the Velleda¹⁰⁸

And I – weak, languid, obscene, digesting, tossing about dismal thoughts – I too was *superfluous* (Sartre, 1963, p.182-184).

Here we see a person suddenly confronted by his own contingent existence – his own superfluity – indeed, by the contingent and superfluous nature of everything around him. And it is in this realisation that he experiences overwhelming *nausea*.

With it seemingly inevitable that this character be condemned to a permanently resigned state of malaise and melancholia, Sartre finds a last-minute reprieve for him (and the reader!) at the end of the novel. In the end, having been “a long way from swimming in bliss”, art promised a kind of salvation for our *nauseous* hero. He initially berated the “mugs” who would seek solace in such things:

To think that there are idiots who derive consolation from the fine arts. [...] [T]he concert halls are full to overflowing with humiliated, injured people who close their eyes and try to turn their pale faces into receiving aerials. They imagine that the sounds they receive flow into them, sweet and nourishing, and that their sufferings become music [...]; they think that beauty is compassionate towards them (Sartre, 1963, p.246).

But he is caught off-guard by a jazz tune that is played for him on a gramophone and we see his scorn dismantled:

Now there is this tune on the saxophone. And I am ashamed. A conceited little suffering has just been born, an exemplary suffering. Four notes on the saxophone. They come and go, they seem to say: ‘You must do like us, suffer in strict time.’ Well, yes!

¹⁰⁸ This is a type of moth.

Of course I'd be glad to suffer that way, in strict time, without any complacency, without any self-pity, with an arid purity. But is it my fault [...] if I am superfluous, if the sincerest and driest of my sufferings trails along heavily [...]? (Sartre, 1963, p.247)

He suddenly admits the world of art – paintings, literature, music – is one he wishes to escape into, away from “the world of municipal parks, of *bistros*, of ports” where he exists, merely, “like other people” (Sartre, 1963, p.248). He sees infiniteness in fine works of art as against the finite nature of his own accidental existence. And he sees the artist and the artist's suffering immortalised in the art itself; he feels envy because “they have cleansed themselves of the sin of existing. Not completely, of course – but as much as any man can” (Sartre, 1963, p.251). We immediately see a dramatic shift in his mood:

This idea suddenly bowls me over, because I didn't even hope for that any more. I feel something timidly brushing against me and I dare not move because I am afraid it might go away. Something I didn't know any more: a sort of joy. (Sartre, 1963, p.251)

He is inspired to write a book, conceding that “it wouldn't prevent me from existing or from feeling that I exist” but that one day it would be behind him and “a little of its light would fall over [his] past” (Sartre, 1963, p.252). He hopes that,

through it, I might be able to recall my life without repugnance. [...] And I might succeed – in the past, simply in the past – in accepting myself. (Sartre, 1963, p.252-253)

For Nietzsche, too, art was a 'godsend' in supplanting religion! He asserted that

with the increasing irrelevance of the idea of the Judaeo-Christian God, the [*Urbarmenschen*] are challenged to assume divine prerogatives, among which the most important is that of creating life-affirming moral and life-enhancing aesthetic values (Flynn, 2006, p.40).

He charged their ability to do this with a grave significance:

[M]oral values of nobility and aesthetic values of the beautiful coalesce in the project of making one's life a work of art. This union of the noble and the beautiful can save us from ourselves

[...] that is, from the despair arising out of our realization that the Universe does not care (Flynn, 2006, p.40-41).

Art, then, according to both Sartre and Nietzsche, is a vital anæsthetic for the numbing of this peculiarly human experience, *Nausea*.

In summary, the *nauseous* recognition of our own contingency – our superfluity – is usually about understanding “the sheer fact that we are and that we do not have to be” (Flynn, 2006, p.59). Underscoring our experience of the non-necessity of our existence is not just the obvious fact that had our parents never met ‘we’ would not be here but also our fixation on the distinction between ‘what’ we are and ‘that’ we are at all (Flynn, 2006, p.59).

Absurdity and Revolt

What, then, is that incalculable feeling that deprives the mind of the sleep necessary to life? A world that can be explained even with bad reasons is a familiar world. But, on the other hand, in a universe suddenly divested of illusions and lights, man feels an alien, a stranger. His exile is without remedy since he is deprived of the memory of a lost home or the hope of a promised land. This divorce between man and his life, the actor and his setting, is properly the feeling of absurdity. (Camus, 2004a, p.443)

According to Flynn, it is the questions ‘Why do we exist?’ and ‘Why is there anything at all rather than nothing?’ that the existentialists take most seriously and it is “the fact of our sheer being there” that they grapple with most ardently (Flynn, 2006, p.59). The meaning of life was certainly “the most urgent of questions” for Camus (Camus, 2004a, p.442). Camus’ musings reveal a belief that all we can know is that we exist and that the world exists; “[t]here ends all my knowledge, and the rest is construction”, he says (Camus, 2004a, p.453). Camus agrees with Sartre and Nietzsche that “whatever meaning our world may harbour is created by individuals either alone or in social relations”, viewing this as “the source of our anguish: we

long for meaning conveyed by a Universe that cares but discover only an empty sky” (Flynn, 2006, p.47). And, says Marino,

[i]t is this terrible combination of the human hunger for meaning and the indifference of the universe that casts the formula for Camus’s important and related concepts of the **absurd** and **revolt** [emphasis added] (2004, p.439).

Camus himself described the *Absurd* as “not in man [...] nor in the world, but in their presence together” (Camus, 2004a, p.462), “born of this confrontation between the human need and the unreasonable silence of the world” (Camus, 2004a, p.460).

On the *Absurd*, Sartre would concur with the sentiments of the protagonist in his novel *Nausea* who, in a moment of self-awareness, said:

[W]ithout formulating anything clearly, I understood that I had found the key to Existence, the key to my Nausea, to my own life. In fact, all that I was able to grasp afterwards comes down to this fundamental absurdity (Sartre, 1963, p.185).

Absurdity, as Sartre reveals it, is not something which can be overcome – it is “an essential, ineradicable aspect of the human condition” (Cooper, 1990, p.145). In his opinion, made abundantly clear, “Man is a useless passion” (Sartre, 1958, p.615).

Enter: existential solace.

In opposition to the more pessimistic outlook assigned to ‘man’ by Sartre, Camus challenged us to make the most of our *absurd* plight. He brought us the example of Sisyphus who “has risen above his fate, not by dull resignation but by *deliberate choice*” (Flynn, 2006, p.48). Camus uses his essay, *The Myth of Sisyphus*¹⁰⁹, as an analogy for the human situation, which he suggests is “pointless and absurd [and unable to be justified] either in religious or humanitarian terms” (Charlesworth, 1975, p.2). And he counsels that “it is in the recognition and willed acceptance of his absurd fate that man transcends his fate” (Charlesworth, 1975, p.2).

¹⁰⁹ First published in 1942.

Camus recounts the story of Sisyphus thus:

The gods had condemned Sisyphus to ceaselessly rolling a rock to the top of a mountain, whence the stone would fall back of its own weight. They had thought with some reason that there is no more dreadful punishment than futile and hopeless labour.

[...]

Sisyphus is the absurd hero. He *is*, as much through his passions as through his torture. His scorn of the gods, his hatred of death, and his passion for life won him that unspeakable penalty in which the whole being is exerted toward accomplishing nothing. This is the price that must be paid for the passions of this earth (2004b, p.489-490).

Camus conjures up an image of Sisyphus “straining to raise the huge stone, to roll it and push it up a slope a hundred times over” and it is on each occasion during the return of the stone “toward that lower world whence he will have to push it up again toward the summit” that Sisyphus is of interest to him (Camus, 2004b, p.490):

That hour like a breathing-space which returns as surely as his suffering, that is the hour of consciousness. At each of those moments when he leaves the heights and gradually sinks toward the lairs of the gods, he is superior to his fate. He is stronger than his rock.

If this myth is tragic, that is because its hero is conscious. Where would his torture be, indeed, if at every step the hope of succeeding upheld him? [...] Sisyphus, proletarian of the gods, powerless and rebellious, knows the whole extent of his wretched condition: it is what he thinks of during his descent. The lucidity that was to constitute his torture at the same time crowns his victory. **There is no fate that cannot be surmounted by scorn** [emphasis added] (Camus, 2004b, p.490-491).

For Marino, it is this last sentence that best articulates the interrelatedness of the concepts of *Absurdity* and *Revolt* (2004, p.439). Camus uses Sisyphus to show that “there is a stoical hope possible to man, even while he recognizes and accepts that life is ‘absurd’” (Charlesworth, 1975, p.8):

[Sisyphus] concludes that all is well. This universe henceforth without a master seems to him neither sterile nor futile. Each atom of that stone, each mineral flake of that night-filled mountain, in itself forms a world. The struggle itself toward the heights is enough to fill a man's heart. One must imagine Sisyphus happy (Camus, 2004b, p.492).

According to Camus, there are three consequences stemming from my awareness of the *Absurd*: "my revolt, my freedom, and my passion" (Camus, 2004a, p.487). One could say that Sisyphus responds to his consciousness of the *Absurdity* of his existence by finding 'micro-meaning' within a 'macro-meaninglessness'. He is able to do this because he realises that

completely turned toward death ([...] the most obvious absurdity), the absurd man feels released from everything outside that passionate attention crystallizing in him. He enjoys a freedom with regard to common rules (Camus, 2004a, p.483). [...] [I]f the absurd cancels all [his] chances of eternal freedom, it restores and magnifies, on the other hand, [his] freedom of action [in the here-and-now]. (Camus, 2004a, p.481)

Sisyphus is completely free to make of his existence what he will.

Camus, on his journey "of finding out whether or not life had to have a meaning to be lived", eventually decides that "it will be lived all the better if it has no meaning [and that] [l]iving an experience, a particular fate, is accepting it fully" (Camus, 2004a, p.479). In a sense, our only hope is in acknowledging that there is no ultimate hope; in choosing to accept this and continuing to live – albeit with limited expectations in view of our mortality – we enact the supreme act of *Revolt* (Flynn, 2006, p.48). Camus tells us that *Revolt* is

[o]ne of the only coherent philosophical positions [...] It is a constant confrontation between man and his own obscurity. It is an insistence upon an impossible transparency. It challenges the world anew every second. [...] It is that constant presence of man in his own eyes. It is not aspiration, for it is devoid of hope. That revolt is the certainty of a crushing fate, without the resignation that ought to accompany it (2004a, p.479).

In a similar vein, Nietzsche advocates “a heroic atheism by which one forges ahead like Sisyphus despite the presumed indifference of the Universe” in the wake of the death of God (Flynn, 2006, p.54). Nietzsche approved of the kind of *Revolt* demonstrated by Sisyphus for having, in his words, “turned the ‘it was’ (his past, the givens of his situation) into the ‘thus I willed it’” (Flynn, 2006, p.48).

So we see that Camus and Nietzsche are convinced that we can ‘overcome’ life’s *Absurdity*. Likewise, de Beauvoir also thinks it may be overcome and suggests that

we can ‘escape the absurdity of [contingency] by escaping the absurdity of the pure moment’. This is done through incorporating each ‘moment’ of life within ‘the unity of the project’ of a whole life (Cooper, 1990, p.144-145).

Cooper disagrees with de Beauvoir on this, saying that few lives possess that “tight structure and integrity” where each “moment” actually “[takes] its place in a carefully executed life-plan” (1990, p.145). And because human existence is riddled with the *Absurd*, he says we know “there is no final, rational determination of the large decisions in life, of our ‘fundamental projects’” anyway (Cooper, 1990, p.149). However, one could say – and I think the existentialists would largely agree – that we have the choice to make of our life a project that consists of achieving existential individuality.

Ambiguity

[T]o say that [existence] is ambiguous is to assert that its meaning is never fixed, that it must be constantly won [...] [I]t is because man’s condition is ambiguous that he seeks, through failure and outrageousness, to save his existence (de Beauvoir, 2004, p.413).

Simone de Beauvoir said that Existentialism has, from the beginning, defined itself as a philosophy of *Ambiguity* (Flynn, 2006, p.66). The existentialists “prize” this *Ambiguity* (Flynn, 2006, p.37) and call for us to face up to the *Ambiguity* of our

existence; we should recognise the many poles between which our existence stands, allowing each to assert itself (Cooper, 1990, p.119). To resolve the *Ambiguity* by ignoring or denying some of the poles of our existence – as with Sartre’s waiter in the café – is to live in *Bad Faith* (Cooper, 1990, p.109). One tension that arises within human life is because people participate in a “public, social world where [they are] the object of ‘the Look’, judgements and categorizations of others” but they are also free individually to make their own meanings from their direct experiences of the world, and people and events around them (Cooper, 1990, p.109). This tension is one of many that render human existence *ambiguous*.

Another tension within human existence is that, as humans, we “exist in-situation” (Flynn, 2006, p.65). According to existentialists, this means that not only are we situated in a body (we are embodied beings) but we also

are an integral part of that universe and the cultural world that envelops it. Less than angels, we are more than machines. **Situation is an ambiguous mixture of what Sartre calls our ‘facticity’ and our ‘transcendence’.** ‘Facticity’ denotes the givens of our situation such as our race and nationality, our talents and limitations, the others with whom we deal as well as our previous choices. ‘Transcendence’ or the reach that our consciousness extends beyond these givens, denotes the takens of our situation, namely how we face up to this facticity [emphasis added] (Flynn, 2006, p.65-66).

Our situation is *ambiguous* in the sense that “one cannot measure off the precise contribution of what is **given** and what is **taken** in each situation” [emphasis added] (Flynn, 2006, p.66). We catch Sartre’s man-at-the-keyhole contemplating this just before he discovers that someone is looking at him:

[My situation] reflects to me at once both my facticity and my freedom; on the occasion of a certain objective structure of the world which surrounds me, it refers my freedom to me in the form of tasks to be freely done. There is no constraint here since my freedom eats into my possibles and since correlatively the potentialities of the world indicate and offer only themselves.

Moreover I can not (*sic*) truly define myself as *being* in a situation [...] since I am what I am not and since I am not what I am—I can not (*sic*) even define myself as truly *being* in the process of listening at doors. I escape this provisional definition of myself by means of all my transcendence. [...] Thus not only am I unable to *know* myself, but my very being escapes—although I *am* that very escape from my being—and I am absolutely nothing (Sartre, 1958, p.259-260).

Our transcendence is wherein all our power lies.

This duality of the human condition as both facticity (our antecedent condition) and transcendence (our possibility) is what allows the possibility of living in *Bad Faith*. Flynn outlines two forms of Sartre's *Bad Faith* that rely on this – “[t]he more common form tries to collapse our transcendence into our facticity” (Flynn, 2006, p.72). One example of this is the person who “flees responsibility by claiming: ‘That’s just the way I am.’”; another is “the attitude of bad faith which allows another subject to determine the ‘identity’ to which we try to conform” (e.g., Sartre’s role-playing waiter) (Flynn, 2006, p.72/73). The other less common attitude of *Bad Faith* is summarised by Flynn as that when we “[discount] our antecedent condition in sheer wishfulness, as if we were pure possibility with no actuality, living entirely in the future, unencumbered by any past” (Flynn, 2006, p.74). In both cases, the *Bad Faith* depends on a lie about the human condition, “insisting that it is either transcendence or facticity when, in fact, it is both but in an ambiguous mix that those who cannot bear to live in ambiguity find unnerving” (Flynn, 2006, p.74). Accepting the *Ambiguity* of this aspect of the human condition and living this truth is part of being an *authentic* individual.

Ambiguity also extends to the realm of moral decision-making. Flynn points out Aristotle’s early warning in this regard, that “it is a mistake to seek a greater degree of clarity than the subject matter allows. You don’t look for mathematical precision in moral matters” (2006, p.66). Sartre’s assertion that there is no God, and without a divine plan therefore no universal moral law, paved the way to a radical view of *Freedom*. A case for normative ethics consisting of moral absolutes could no longer

be made because it was seen that there can be no rational basis for claiming that one action is any better than another (Marino, 2004, p.412). Instead, said de Beauvoir, we can be counselled on **how to make** moral choices: "Ethics does not furnish recipes any more than do science and art. One can merely propose methods" (2004, p.417). In light of this – with ethics no longer able to provide absolute direction for our decision-making – Simone de Beauvoir decreed that human beings are condemned to *Ambiguity*.

Freedom and Responsibility

Existentialism is a philosophy of freedom. Its basis is the fact that we can stand back from our lives and reflect on what we have been doing. In this sense, we are always 'more' than ourselves. But we are as responsible as we are free (Flynn, 2006, p.8).

Sartre announced that "man is condemned to be free" (Sartre, 1975, p.353) – a seemingly curious statement given that 'condemnation' implies a kind of damnation, and yet *Freedom* is commonly acknowledged as the supreme value of existentialist thought (Flynn, 2006, p.xi). What could Sartre have meant by this?

He, as with many existentialists, was deeply influenced by Nietzsche's revelation that God is dead. As was stated earlier, both Sartre and Camus believed

the rejection of God, and especially of the God of Christianity, is necessary if man is to take himself and his freedom seriously. Both of them think that religion alienates a man from his true self and provides him with a way of evading the full consequences of his freedom (Charlesworth, 1975, p.17).

Existentialism, on the other hand, is "nothing else but an attempt to draw the full conclusions from a consistently atheistic position" (Sartre, 1975, p.369). A wonderfully concise section of Sartre's *Existentialism is a Humanism*, in which he discusses our *Forlornness* and our *Freedom*, neatly summarises what he means:

When we speak of **forlornness**, a term Heidegger was fond of, we mean only that God does not exist and that we have to face all the consequences of this. The existentialist is strongly opposed to a certain kind of secular ethics which would like to abolish God with the least possible expense. [...] The existentialist [...] thinks it very distressing that God does not exist, because all possibility of finding values in a heaven of ideas disappears along with Him; there can no longer be an *a priori* Good, since there is no infinite and perfect consciousness to think it. Nowhere is it written that the Good exists, that we must be honest, that we must not lie; because the fact is we are on a plane where there are only men. Dostoevsky said, **“If God didn’t exist, everything would be possible.”** **That is the very starting point of existentialism.** Indeed, everything is permissible if God does not exist, and as a result man is forlorn, because neither within him nor without does he find anything to cling to. He can’t start making excuses for himself.

If existence really does precede essence, there is no explaining things away by reference to a fixed and given human nature. In other words, **there is no determinism, man is free, man is freedom.** [...] **We are alone, with no excuses.**

That is the idea I shall try to convey when I say that **man is condemned to be free. Condemned, because he did not create himself, yet, in other respects is free; because, once thrown into the world, he is responsible for everything he does.** [all emphases added] (Sartre, 2004, p.348-350)¹¹⁰

Sartre rejected the notion of the Freudian unconscious, denying the potency of the kinds of drives and forces that supposedly rob us of our *Freedom and Responsibility* (Flynn, 2006, p.49). In doing so, he famously maintained that “man is responsible for his passion” (Sartre, 2004, p.350). The defining characteristic of Sartrean Existentialism, which he refers to in the passage above, is his claim that **existence precedes essence**. By this he means to say that “what you are (your essence) is the result of your choices (your existence) rather than the reverse. Essence is not

¹¹⁰ This contains an alternative English translation of *L’existentialisme est un humanisme* to that published in 1975 in the book edited by Walter Kaufman. I have cited from both texts in this thesis – in each instance choosing whichever I felt was the most articulate translation into English.

destiny. You are what you make yourself to be" (Flynn, 2006, p.8). In adding to this by then saying, "We are alone, with no excuses", he follows on from

the Nietzschean view that, in the absence of belief in God, we should assume the absolute freedom that Descartes had ascribed to the Divinity. In phenomenological terms, this meant that the entire 'world' (the horizon of our meanings) is our creation for which we hold total responsibility (Flynn, 2006, p.43).

And so, remaining in the aftermath of Sartre was an atheistic world where:

individuals were left to create their own values because there was no moral order in the universe by which they could guide their actions, indeed, that this freedom was itself the ultimate value to which one could appeal (as [Sartre] put it, 'in choosing anything at all, I first of all choose freedom') (Flynn, 2006, p.45).

In fact, Sartre and de Beauvoir saw *Freedom* itself as the defining feature of 'man' (Sartre, interview with Charlesworth, 1975, p.26).

Michel Onfray, a contemporary French philosopher and author of *The Atheist Manifesto*, says that too many of us who do not believe in God do not assume the 'absolute freedom' which flows on from this, **continuing to live instead as if there were a God**: "[M]any individuals who consider themselves atheists profess—without noticing it—an ethic, a way of thinking, a vision of the world saturated in Judeo-Christianity" (Onfray, 2007, p.45). However, in the Western world, escaping the shackles of Judeo-Christianity is no trivial task given his assertion that even our bodies are imbued with Christianity:

Two thousand years of Christian discourse—atomy, medicine, physiology, of course, but also philosophy, theology, and aesthetics—have fashioned the body we inhabit. And along with that discourse we have inherited Platonic-Christian models that mediate our perception of the body, the symbolic value of the body's organs, and their hierarchically ordered functions. [...] We accept the spiritualization and de-materialization of the soul, the interaction of sin-prone matter and of luminous mind [...] All have contributed to Christianity's sculpting of the flesh (Onfray, 2007, p.47).

But while the existentialist philosophers acknowledge that “[w]e are born biological beings” (and then, according to Onfray, socialised into becoming Christian beings), they insist that

we must become existential individuals by accepting responsibility for our actions. This is an application of Nietzsche’s advice to ‘become what you are’. Many people never do acknowledge such responsibility but rather flee their existential individuality into the comfort of the faceless crowd (Flynn, 2006, p.x)

Becoming an existential individual – living *authentically* – is about facing up to the fact that we are radically *free*. We will never achieve this so long as we shirk the *Responsibility* this brings by entering into the secure embrace of the various forms of *Bad Faith* (Cooper, 1990, p.148).

So how is it that *Freedom* brings with it *Responsibility*? It begins with the fact that we can pause to reflect on our lives –

we are always more than ourselves and hence without excuse. In other words, our temporalizing consciousness of what we are is always enough ahead of what we are that Sartre can claim that whatever we may be, we are in the manner of ‘not-being’ it. It is this gap which temporalizing consciousness introduces into our lives that accounts for our freedom and grounds our responsibility (Flynn, 2006, p.70).

Nietzsche’s *On the Genealogy of Morals* contains his account of “the long story of how *responsibility* originated” (2004, p.147). According to this, the highest attainment of an evolving civilisation – “the end of [a] tremendous process” – is to produce a society in which ‘man’ is to become the “*sovereign individual*” – liberated from “morality of custom, autonomous and supramoral (for ‘autonomous’ and ‘moral’ are mutually exclusive), [...] [a] man who has his own independent, protracted will and the *right to make promises*” (2004, p.147-148). Nietzsche describes this person as an “emancipated individual, with the actual *right* to make promises, this master of a *free will*” – his “mastery over himself also necessarily gives him mastery over circumstances, over nature, and over all more short-willed

and unreliable creatures" (2004, p.148). He goes on to say that 'men' who are 'free', who possess

a protracted and unbreakable will [...] give their word as something that can be relied on because they know themselves strong enough to maintain it in the face of accidents, even "in the face of fate". [...] The proud awareness of the extraordinary privilege of *responsibility*, the consciousness of this rare freedom, this power over oneself and over fate, has in his case penetrated to the profoundest depths and become instinct, the dominating instinct. What will he call this dominating instinct, supposing he feels the need to give it a name? The answer is beyond doubt: this sovereign man calls it his *conscience* (Nietzsche, 2004, p.148).

The sovereign individual has 'a conscience', as we say, because they are acutely aware that mastery over their own will implies *Responsibility* for their actions and choices.

In *On the Genealogy of Morals*, Nietzsche talks of punishment, the concept of justice, and *Freedom*. He says it was a fear of medieval punishments which truly gave rise to the origin of guilt and personal obligation for "reason, seriousness, mastery over the affects, the whole somber (*sic*) thing called reflection [...] [and] the other 'somber thing,' the consciousness of guilt", he claims, were all "bought" with "much blood and cruelty" (2004, p.151). Furthermore, he considers our *Freedom* to be constrained because we live indebted to our community for all the protections and advantages that a "communality" affords us – and when we accede to living in a society where "one dwells protected, cared for, in peace and trustfulness, without fear of certain injuries and hostile acts", we bind and pledge ourselves to obeying its laws (Nietzsche, 2004, p.160). According to Nietzsche, then, it is this indebtedness and fear of punishment from whence our *Responsibility* for others truly originated, rather than it being due to some kind of moral imperative or notion of 'goodness'.

Some see the *Existential Freedom* and concomitant *Responsibility* inherent in the human condition as too overwhelming for those immersed in the 'they' to bear. Such is the burden on the individual that Sartre says, "every man, without any

support or help whatever, is condemned at every instant to invent man" (1975, p.353-354). Elsewhere, in *Being and Nothingness*, he comments that "there are no accidents in a life" because "[e]verything which happens to me is *mine*" (1958, p.554) – no matter what happens to me, my reaction to it and how it shapes me will teach me something about myself rather than the event as such:

[M]an being condemned to be free carries the weight of the whole world on his shoulders [...] [T]he responsibility of the for-itself is overwhelming since he is the one by whom it happens that *there* is a world; since he is also the one who makes himself be, then whatever may be the situation in which he finds himself, the for-itself must wholly assume this situation with its peculiar coefficient of adversity¹¹¹ [...] He must assume the situation with the proud consciousness of being the author of it, for the very worst disadvantages or the worst threats which can endanger my person have meaning only in and through my project [...] It is therefore senseless to think of complaining since nothing foreign has decided what we feel, what we live, or what we are (Sartre, 1958, p.553-554).

The emphasis on individualisation here "makes you or me, not man in general, 'the maker of a world'" and, by doing this, Sartre makes it plain to see "the radical responsibility each of us bears" (Cooper, 1990, p.147). Cooper says that our *Responsibility* extends even to our character because this emerges out of our attitudes and values and these are open to examination and revision (1990, p.150).

The idea that radical *Freedom* and *Responsibility* are too much for the 'they' to confront and cope with is prevalent in existential literature. A reading of Dostoevsky's *The Brothers Karamazov* (in particular, the section on 'The Grand Inquisitor') (Dostoevsky, 2004) and Unamuno's *Saint Manuel Bueno, Martyr* (de Unamuno y Jugo, 2004) gives an insight into those who would 'protect' the 'they' by encouraging belief in God so they do not live in a perpetual state of *Existential Angst*. They seem to be saying, 'Take pity on them! Do not open their eyes to the

¹¹¹ What Sartre means by 'coefficient of adversity' here may be better understood in the following quote: "the situation is *mine* because it is the image of my free choice of myself, and everything which it presents to me is *mine* in that this represents me and symbolizes me. Is it not I who decide the coefficient of adversity in things and even their unpredictability by deciding myself?" (Sartre, 1958, p.554)

meaningless of Being (the Nothingness which skirts around Being) and its necessary co-conspirators, *Fear*, *Forlornness* and *Freedom*, as these can bring only *Anguish* and overwhelming *Responsibility*.'. Both of these stories speak to Nietzsche's doctrine of the *Übermenschen* – that there are only a few culturally elite "choice souls [...] who are capable of making their own values for themselves, of bearing the burden of the death of God" (Charlesworth, 1975, in his interview with Carroll, p.20). The rest, when fully realising the sheer scope of their *Freedom* and the *Responsibility* this brings, suppress it through the devices of *Bad Faith* (Cooper, 1990, p.148). In this way though, *Bad Faith* can be seen as a mechanism that the 'they' use to cope with the terrific burden that *Responsibility* and *Anguish* bring. And while this is not living *authentically*, it is in fact a rebuttal of the concerns held by those who would see themselves as necessarily 'protecting' the 'they' by keeping them 'in the dark'.

The existentialists would view this paternalistic mentality as unconscionable and unjustifiable – these people are being kept by others from achieving their ownmost possibilities of being. In this oppression, Sartre would say that the oppressors oppress themselves (Cooper, 1990, p.181) because he believed that "I am obliged to will the liberty of others at the same time as my own" (Sartre, 1975, p.366). Cooper explains what he thinks Sartre meant here: "Through treating others as alien, I become alienated from myself, and my freedom becomes an 'oppressed freedom' through my effective denial of others' freedom" (Cooper, 1990, p.181). Finally, Sartre said "I can form judgments upon those who seek to hide from themselves the wholly voluntary nature of their existence and its complete freedom" (1975, p.366) but people must be given the chance to choose for themselves – and, if this is the case, the existentialists must especially condemn those who deliberately seek to withhold this choice from others.

Freedom and Choice

*Because we are fundamentally in-situation, and because this situation is as flowing and ambiguous as are time and consciousness themselves, humans are not stable, timeless identities. [...] We are fundamentally a work in progress, a story in the process of being written*¹¹² (Flynn, 2006, p.69).

It was Kierkegaard who said that, as human beings, our situation is such that the choices we make are self-constituting and, because the “either/or” nature of choosing “involves risk, commitment, and individuation”, our choices are also liberating (Flynn, 2006, p.32). Sartre endorsed Kierkegaard’s relation of choice to self-constitution, adding that “for human reality, to be is to choose oneself” (Flynn, 2006, p.33). He concurred that choice is individuating, as evidenced by his famous advice to a pupil of his who was unsure which of two courses of action available to him was the ‘right’ one for him to pursue:

You are free, therefore choose—that is to say, invent. No rule of general morality can show you what you ought to do: no signs are vouchsafed in this world (Sartre, 1975, p.356).

He explains that it is left to the individual alone to interpret these signs (to choose their meaning) and, for their ‘decipherment’, therefore, “he bears the entire responsibility. That is what ‘abandonment’¹¹³ implies, that we ourselves decide our being. And with this abandonment goes anguish” (Sartre, 1975, p.357). The *Anguish* of which Sartre speaks here is the sense of “complete and profound (*sic*) responsibility” which belongs with a person’s “fully realizing that he is not only choosing what he will be, but is thereby at the same time a legislator deciding for the whole of mankind” (Sartre, 1975, p.351).

¹¹² Denial of this condition places a person in *Bad Faith* because, as stated previously (see section: *Bad Faith* and the ‘*Predominance of the Other*’), they do not remain “open to the transformative effect that the possibility of future choices and future actions could bring [and hence] they have lost their freedom” (Cooper, 1990, p.117).

¹¹³ This word, ‘abandonment’, is substituted with ‘forlornness’ in alternative translations of this text (i.e., Sartre, 2004).

Nietzsche's views on *Existential Freedom* and its relationship to the choices we make differed somewhat from Sartre's. Nietzsche famously denied the notion of free will and the moral choice that it exercises (Flynn, 2006, p.37). In Flynn's words, Nietzsche believed that "[t]he 'error' of free will [...] is the belief that choice rather than physiological and cultural forces is the basis of our judgements of moral approval and disapproval" (2006, p.38). So, in looking for "[grounds] for the responsibility that we feel in ourselves and ascribe to others" (in the way that existentialists construct the enduring problem of freedom versus determinism) (Flynn, 2006, p.38), Sartre claims that people are absolutely *free* to behave as they like (free will means they have absolute control over their behaviour) whereas Nietzsche says that despite being *free*, people cannot be in absolute control of all their irrational instincts and drives. Rather than free will, he said the real reasons for why we do what we do are attributable to his concept of '**will-to-power**' (understood psychologically, this is what he called the innate drive to dominate and control) and yet he saw "the self-control exercised by the free spirits for whom [he] reserves a 'higher' morality than the chiefly religious ethics of the herd" as the "'highest' expression" of this will-to-power (Flynn, 2006, p.38).

In Cooper's estimation (1990, p.153), Merleau-Ponty gives the most considered statement of *Existential Freedom* in *Phenomenology of Perception* (1962), where he criticises Sartre's account of *Freedom* and articulates a version that he sees as more harmonious with the tenets of existential phenomenology. Cooper (1990, p.151) saw both this and Heidegger's account of *Freedom* as superior to the more simplistic account offered by Sartre. One of Merleau-Ponty's criticisms of *Sartrean Freedom* was that:

I am situated in a social environment, and my freedom, though it may have the power to commit me elsewhere, has not the power to transform me instantaneously into what I decide to be (1962, p.447).

Deciding upon and then becoming who I want to be requires not only that I choose what that is but that I come to be identified as that "through an implicit or

existential project which merges into [my] way of patterning the world and co-existing with other people" (Merleau-Ponty, 1962, p.447).

Cooper explains this critique of Sartre thus: while a person may have the **will** to change their belief, they cannot "'annul' [their] stance towards the world" instantaneously because they must acquire new understanding, thereby becoming immersed in a new way of seeing; until this occurs, "there can be no belief, wilful or not" (1990, p.154). And so, it is not so easy to 'not-be' who we are, as Sartre would put it. It cannot happen as the result of a momentary decision because the framework of interpretation and belief that we 'choose' presupposes the acquisition of understanding (Cooper, 1990, p.154). Much of the understanding we acquire in the early part of our lives is the result of information spoon-fed to us – we are fairly passive, uncritical recipients of what we are told as children and many in society continue to be so in terms of what they 'learn' from the media and other sources of information around them. So, following on from Merleau-Ponty's point, there can be no 'initial choice'¹¹⁴ as to the framework of interpretation and belief that we start out with in life (as Sartre propounded) – this being largely the result of the 'thrown'¹¹⁵ character of our existence, as Heidegger said (Cooper, 1990, p.153-154). And while 'modifications' to it can occur, these are made gradually over time rather than at one fell swoop (Cooper, 1990, p.154). What this means is that on any occasion when we are presented with a choice, how truly *free* we are in making this choice is limited by: 1) our 'thrownness' in an already interpreted, public world; and 2) previous choices we have made (**what has been** our situation) – but on each occasion, and over each situation that we have a choice, we remain *free* to choose as we will.

¹¹⁴ 'Initial choice' is "supposed to give rise to all understanding, to bring it about that *there is* a world for a person" (Cooper, 1990, p.154). According to Cooper (1990, p.153), de Beauvoir (1948) saw the 'initial choice' to be a "pure contingency", while Sartre (1957) maintained that it is something one does freely.

¹¹⁵ The word 'thrownness' means "immersion at the outset in a world already interpreted by one's fellows, a world a person must first be 'in' before he can make an 'issue' of it and of himself" (Cooper, 1990, p.154).

But, to Merleau-Ponty, the demise of 'initial choice' does not imply the diminution of *Existential Freedom* for he (and Marcel too) said that Sartre should never have tied *Freedom* and Choice together in the first place (Cooper, 1990, p.155). He said:

I can no longer pretend to be a nihilation (*néant*), and to choose myself continually out of nothing at all. [...] I am a general refusal to be anything, accompanied surreptitiously by a continual acceptance of such and such a qualified form of being. [...] I can at any moment interrupt my projects [because I have] the power to begin something else (Merleau-Ponty, 1962, p.452).

We have powers – “the power of ‘general refusal’ and the power to ‘begin something else’”¹¹⁶ – and these are what constitute *Existential Freedom*, “for it is they, exercised or not, which make a person responsible for his stance towards the world and for the actions which arise from it” (Cooper, 1990, p.155).

Merleau-Ponty went on to say that non-refusal should not be confused with Choice in the way of Sartre in his earlier work:

We must not say that I continually choose myself, on the excuse that I *might* continually refuse what I am. Not to refuse is not the same thing as to choose. (1962, p.452)

In support of Merleau-Ponty's words, Cooper says the average person is not

permanently in the process of considering and selecting competing interpretations and values. In reality, we 'natural selves' are normally embroiled in the business of living in a world whose contours are not experienced as products of will. (1990, p.155)

People often act unthinkingly without constantly reviewing or modifying the path they are taking, particularly when it comes to negotiating the minutiae of their everyday existence.

In view of this critique of *Sartrean Freedom*, Cooper prefers Heidegger's take on *Authenticity* – that we **win** it from thrown existence in the 'they', rather than **lose** it in the Sartrean way (Cooper, 1990, p.154-155). This, then, is a kind of “reactive

¹¹⁶ Here, Cooper cites Merleau-Ponty (1962).

freedom”, which for Heidegger’s *Dasein* is “a freedom ‘released from the illusions of the ‘they’, yet ‘within the limitations of its thrownness’”¹¹⁷; for Merleau-Ponty it is about the “power to refuse”; and for Martin Buber it is “an ‘open confrontation’ with sedimented ways that have become ‘adverse’ to the emergence of new meanings”¹¹⁸ (Cooper, 1990, p.156). Cooper goes on to explain that it is this reactive kind of *Existential Freedom* that eventually filtered through and was favoured by the ‘existentialist philosophers’ of more recent years. An example he provides of this, a snapshot of Charles Taylor’s words, is well-chosen: the responsibility for oneself is “not that of radical choice, but [of] radical evaluation”¹¹⁹ – “‘radical evaluation’ of one’s desires and values in our concept of what it is to be a person” (Cooper, 1990, p.156/159). Again, that power to begin something else...

Care and the Technological Attitude

[T]echnology, in the normal sense of the word, is just one small part of the technological attitude, an attitude which arises as a result of seeing humanity as the center of the universe (Lemay & Pitts, 1994, p.76).

Heidegger used the term, *Technology*, to describe a particular way of existing in the world; a specific way of seeing the world. In the *Technological Attitude*, everything is revealed to us in a certain way – we position ourselves as the “thinking thing” against all other beings, which are seen as something there for us to consume; as “stuff” which exists exclusively for our purposes (Lemay & Pitts, 1994, p.71/72). Heidegger called this *Technological Attitude* ‘*gestell*’¹²⁰ because it divides up “stuff” (or “stock”)¹²¹, thus enframing it for our use. He saw this as the prime reason behind humanity’s “abuses of nature”:

¹¹⁷ Here, Cooper paraphrases Heidegger (1962).

¹¹⁸ Here, Cooper paraphrases Buber (1937).

¹¹⁹ Cooper cites Taylor (1982).

¹²⁰ This word translates as ‘framing’ in English.

¹²¹ Heidegger used the word ‘*bestand*’ to describe beings once they have been transformed by technology into ‘stuff’. This word translates as ‘standing reserve’ or ‘stock’ in English.

If the world exists because of humanity, then there is nothing humanity cannot do to it or with it. [...] The world exists to be used. It exists for those thinking things who have the power to exploit it. (Lemay & Pitts, 1994, p.78-79)

Heidegger was convinced that we will only begin to live in harmony with the rest of the world when we see “that all the beings in the world are interconnected, and that humanity is just one of these beings” (Lemay & Pitts, 1994, p.83). Art, he says, helps us to do this because it “is a way of appreciating the interconnections among beings that technology ignores” (Lemay & Pitts, 1994, p.83). This alternative to the *Technological Attitude* he called *Care*, and it is only through this way of looking at the world that he believed the possibility opens up before us to live *authentically* – to make the most of our own possibilities (Lemay & Pitts, 1994, p.82/58).

From the existentialist viewpoint, our ability to reason and understand is reliant on our passions, our desires, our moods, our feelings (Cooper, 1990, p.87). In talking about this, Cooper also elaborated on all that is encapsulated in the Heideggerian concept of *Care*:

The world is ‘proximally’ understood as one ‘lit up’ in relation to our purposive activities. But these are the purposes of creatures with desires and concerns, who experience the joys and frustrations consequent upon the outcomes of their activities. Beings who only stand and wait, stop and stare, would have no world to understand and reason about. [...]

[...] [N]either understanding nor desire and feeling are to be construed as mere accompaniments to our activities. These activities are not the visible products of distinct inner processes, but the vehicles of a ‘caring’ engagement with the world. It is only permissible to decompose that engagement into bodily, cognitive and affective components when it is appreciated how artificial and provisional this procedure is. (1990, p.87)

Heidegger’s take on the human condition, as one essentially defined by *Care*, has been summarised “as fundamentally a matter of being fated to a self and to a world of other selves and objects about which one cannot choose not to be concerned” (Mulhall, 1996, p.112). Further to this, Mulhall explained that Heidegger’s point is

not that Dasein is always caring and concerned, or that failures of sympathy are impossible or to be discouraged; it is rather that, as Being-in-the-world, Dasein must *deal* with that world. The world and everything in it is something that cannot fail to matter to it. (1996, p.111)

According to Cooper, this means our fundamental relation to the world manifests not as a relation “of substances causally interacting with others” but as a relation to things “in so far as they matter to us for the ‘issue’ that each of us is to himself” (Cooper, 1990, p.157). In other words – and this being “[t]he central proposition of existential phenomenology” – “we exist in a ‘human world’ whose contents are articulated in terms of the significance they have through the intentional projects in which we engage” (Cooper, 1990, p.157). More simply put (perhaps), the significance of objects, events and other beings to us as individuals will depend on what is important to us in life and what we ‘want out of life’ – that is, as we view them against our life ‘project’ or ‘self-defining Choice’¹²².

Merleau-Ponty wrote on this under the title of ‘Freedom’ in his *Phenomenology of Perception*:

Even what are called obstacles to freedom are in reality deployed by it. An unclimbable rock face, a large or small, vertical or slanting rock, are things which have no meaning for anyone who is not intending to surmount them, for a subject whose projects do not carve out such determinate forms from the uniform mass of the *in itself* and cause an orientated world to arise—a significance in things. There is, then, ultimately nothing that can set limits to freedom, except those limits that freedom itself has set in the form of its various initiatives, so that the subject has simply the external world that he gives himself. Since it is the latter who, on coming into being, brings to light significance and value in things, and since no thing can impinge upon it except through acquiring, thanks to it, significance and value, there is no action of things on the subject, but merely a signification (1962, p.436).

¹²² The notion of the ‘self-defining Choice’ is one put forward by Sartre and it is this “that brings the multiplicity of our concerns into a whole and invites our authentic embrace” – this is the unifying meaning and direction of our lives (Flynn, 2006, p.76).

As self-defining subjects, and notwithstanding the influence of others, it is ultimately we alone who confer upon objects, events and other beings their significance and value – their meaning (to us).

SUMMARY

Preliminary analysis of the emerging data from the first interviews in this study found it to be replete with the dilemmas and paradoxes of human existence. I believed the data pointed wholeheartedly to existential philosophy as a fitting explanatory device. In response to this, I saw a need to devise a method of understanding based on the concepts that pervade the existentialist literature. This section represents the fulfilment of my goal to formulate such a 'method'. I have restricted the concepts summarised here to those that I saw were pertinent to the data arising in the context of this study. These existential concepts – *Despair, Self-estrangement and Authenticity, Bad Faith and the Predominance of the Other, The Look, Fear and Anguish (Angst), Nausea, Absurdity and Revolt, Ambiguity, Freedom and Responsibility/Choice, and Care and the Technological Attitude* – were all later examined in relation to, and as they defined, the topics uncovered and discussed in the interviews with the participants. They became an interpretive lens through which I was able to offer a deepened analysis of this data. In turn, the data itself validated and became illustrative of the existential concepts as I have delineated them. I also claim that an existentialist 'ethic', based on the foundation of the willing of *Freedom* for oneself and all others, can be used as a tool by which to judge the decisions and values that are enacted in the genetic counselling/testing context. This ethic will be developed in the latter part of Chapter Four.

CHAPTER FOUR: METHOD

RESEARCH METHOD AND DESIGN CONSIDERATIONS

This study uses concepts derived from existential philosophy to provide an understanding of the hopes and concerns of parents within 'carrier families'. A qualitative data collection method was selected after extensive philosophical consideration as it was believed to be the most appropriate and ethically responsible approach to answering the research question: "What is it like to be the parent of a child who is, or could be, a carrier of cystic fibrosis?".

Minichiello *et al.* have said that qualitative (or post-positivist) approaches to research "allow for a recursive uncovering and discovery of meaning and the essence of human experience" and this is why they attract the researcher who is interested in "the meanings invested in life and action by human beings, and in the narratives which they construct to develop and support individual life meaning" (1995, p.129). A qualitative research approach is less "geared to finding out the truth *per se* [than it is to finding out] the truth as the informant sees it to be" (Minichiello *et al.*, 1995, p.94), this being compatible with my interest in the second-hand account of the informants in this study. The in-depth interview method was chosen because it is thought to be "one of the more effective means of gaining access to that account" (Minichiello *et al.*, 1995, p.70), allowing an insight into "people's experience of social reality through their routinely constructed interpretations of it" (Minichiello *et al.*, 1995, p.69)

Ethics approval

Ethics approval was obtained from the Social and Behavioural Research Ethics Committee at the university in which I was enrolled when I was to commence the data collection phase of the study (see Appendices 1 & 2). Access to the contact details and medical genetics files of potential participants was negotiated through the research ethics committee of the hospital concerned, and in consultation with the respective Heads of the Clinical Genetics Service and the Chemical Pathology

Unit at the hospital. The hospital's research ethics committee, in giving tentative approval to the research protocol, requested that the planned participant selection process be modified because the proposed method "might bias results" (see Appendix 3). I responded by letter to the Committee's concern (see Appendix 4), explaining that random selection is not necessarily compatible with qualitative research methods and that a biased sample is actually welcomed where the data may be the richer for it, and attached a copy of an editorial from one issue of *Qualitative Health Research* (Morse, 1998) that would substantiate my claims in this regard. The Committee's approval was consequent to this (see Appendix 5)!

Participants

I was interested in families living within a particular state in Australia where at least one child was identified as a carrier of cystic fibrosis before they could give informed consent on their own behalf, or where at least one infant or child is at high risk of being a carrier but has not been tested (i.e., they have a sibling with the disease or they have a parent who is a known carrier). Their known carrier status could have been revealed by:

- 1) a prenatal test for CF (where the disease was ruled out);
- 2) routine neonate screening for CF (a false positive result in the first tier of the screening process for the disease); or
- 3) a genetic test during childhood at the parents' request.

The study participants were the mothers and/or fathers in these families (two grandparents were also interviewed).

Potential participants who fit the study criteria were identified from medical genetics files stored in the Clinical Genetics Service and Chemical Pathology Unit at a major metropolitan children's hospital in Australia. The contact details recorded in the files were either confirmed as current or updated by cross-checking them against the listings in the local telephone directory or on the state electoral roll, to which the Clinical Genetics Service has access. In the quest to maintain confidentiality, the names and contact details of individuals and families were

recorded in a separate Excel spreadsheet from any of their (codified) genetic information deemed relevant to this study.

A letter of invitation and accompanying information sheet (see Appendices 6 & 7) were sent out to all potential participants under the name of the Director of the Clinical Genetics Service at the hospital. The letter advised them of the study, the criteria for participation, and invited them to fill out a 'Consent to Be Contacted' Slip for their return to me by post in a reply-paid envelope if they wished to register their interest in participating in the study. In all, letters of invitation were sent out to 77 sets of parents and I received 21 responses (27%) from parents who gave their consent to be contacted directly by me about the study. I was also advised of another five sets of parents who gave their consent to be contacted about the study, having heard about it via word-of-mouth from various clinicians at the hospital.

Sampling

My approach to sampling was compatible with the aim of qualitative research which is to "understand and clarify social processes rather than quantify the distribution of key characteristics within a given population" (Reed, Procter, & Murray, 1996, p.54). Qualitative sampling is geared towards finding information-rich cases, rather than towards randomisation and generalisability. Qualitative researchers seek out those who have a story to share, those who have an unusual or interesting experience to relate, and pounce on the rogues whose stories surprise us and challenge the assumptions that we may take for granted! Qualitative sampling does not claim 'representativeness' but in this study it will be important to sample for variety across the phenomenon of parenting a child who is a known or possible CF carrier. For this reason, I interviewed a mix of parents in terms of: 1) how the parents became aware of their child's CF carrier status; 2) whether their children were yet aware of their carrier status; and 3) whether they had another child with CF, another family member with CF, or did not have any relatives at all with the disease. Eighteen parents/grandparents participated in the study, with thirteen open-ended, in-depth interviews conducted in total.

Biographies of participants¹²³

1 STACY:

Stacy is 37 years old and married to Steve. They have two children who Stacy looks after full-time, while Steve works in the transport industry. They both discovered they were carriers after their first child, Bradley, failed to thrive and was diagnosed with CF at a few months of age based on clinical examination and a sweat test. At this point, the family reminded them that this was the disease that Steve's cousin had died from decades earlier. There had been a lot of family secrecy around his sickness back then. Stacy and Steve decided to conceive another child but chose to have prenatal diagnosis. Their second baby was found not to have CF but the test revealed that she was a carrier. Kristy was born. Bradley is now twelve years old and Kristy is nine years old.

2 HENRY:

Henry is a 73-year-old retired sailor and he is Stacy's father. English is not his first language but he has lived in Australia for decades. His wife passed away almost four years ago and together they had three children: Stacy, another daughter three years later, and a son who is now twenty-five years old. After the birth of Stacy's affected son, the whole family was carrier-tested. It was revealed that Stacy's brother, who was in his early teens at the time, was a carrier but that the other sister was not. To Henry's surprise, it turned out that it was he who carried the less common CF gene mutation that had been passed on to his grandson.

3 ROSE:

Rose is now in her late twenties and was on a working holiday in England eight years ago when her brother's first child, Susie, was born and diagnosed with cystic fibrosis through the neonatal screening program. Six months later in the United Kingdom, Rose fell pregnant; a carrier test at this point revealed that she too was a CF carrier. According to Rose, there was no neonatal screening program for CF in

¹²³ These are all pseudonyms, as are any children's or partners' names referred to in this thesis.

the United Kingdom at that time but she was very insistent that her first son be tested for CF when he was born because the child's father, who she separated from at an early stage of her pregnancy, had not presented for a carrier test when she requested him to. She did not want to undergo amniocentesis because of the miscarriage risk associated with this. She insisted her son be carrier-tested as a baby when it became apparent that he was not affected with the disease; he was identified as a CF carrier. Rose and her new partner had very recently given birth to another healthy son and she planned to initiate having him carrier-tested at his 'six week check'.

ROSE'S MUM:

Rose's Mum had a carrier test but was not shown to be a carrier of the CF gene mutation which had been passed on to Rose and her brother. Instead it was her former husband who carried the faulty copy of the gene.

4 BETTY & DON:

Betty is 46 years old and married to Don. Their youngest daughter, Elise, was identified via ultrasound as having a ruptured bowel one month before she was born and was diagnosed with CF shortly after birth. At this point, they realised this must be the disease that Don's cousin's 22-year-old daughter also has. She too had been born with a ruptured bowel but had only been given two years to live at the time. Elise's two older sisters were eleven and thirteen years old when she was born. Carrier-testing performed two or three weeks after the birth of their affected baby sister revealed that they were both carriers of the disease gene.

5 WENDY:

Wendy is a 32-year-old woman who is the sole carer of her four healthy children, aged 14, 11, 4 and 3 (she lost her third-born child to "cot death"). Wendy grew up in another state as the oldest of three children; however, her younger sister and brother were both affected with cystic fibrosis. Her sister died shortly after turning nineteen when Wendy was twenty-two years old, and her brother died at twenty years of age just a few years later. Wendy had her first son when she was eighteen – the risk that this child would have CF did not occur to her until after he was born.

Her next two boys had a different father who did not seem fazed by the CF risk. Her next partner had a carrier test and was found not to carry any of the common CF mutations. They then had a healthy son and daughter together. To date, none of her children have been carrier-tested.

6 JOSIE:

Josie is a 30-year-old full-time mother who lives with her partner and their three children. She gave birth to twins, a boy and a girl, nine years ago. Three weeks later, the Guthrie test revealed that their baby daughter, Diane, had CF. Neither Josie nor her partner had heard of the disease prior to this. They later decided that they wanted to have another baby but, knowing that they would not terminate an affected foetus, and concerned about the risk of miscarriage associated with chorionic villus sampling and amniocentesis, they chose not to have prenatal testing. They had a baby girl three years after the twins. Blood from her umbilical cord was taken immediately so that a genetic test could be done without the need to await the Guthrie test results – this revealed she was a carrier of one CF mutation.

7 CINDY:

Cindy is a 42-year-old woman, now divorced, who is a pre-school director. She has a healthy son and daughter, aged 17 and 15 respectively. Her parents and her older brother are alive and well. She had a younger brother, born four years after her, who was diagnosed with cystic fibrosis at one year of age after displaying clinical symptoms of the disease. They grew up in a remote area and he was quite well until age six when he began “going downhill” and required a lot of hospital visits. He died when he was nine years old, alone in hospital. Carrier-testing was not available when Cindy had her children and she was not offered amniocentesis. Her risk of having a child with CF was only a passing thought for her when she was pregnant with her first baby. Her husband had no known history of CF in his family and her carrier risk was never an issue for him. She and her older brother were eventually carrier-tested about seven to eight years ago and this confirmed they are both CF carriers. Her children are yet to be tested.

8 ANNE & KEVIN:

Anne is a 42-year-old library manager married to Kevin who is a 44-year-old public servant. They live with their two healthy daughters who are now eleven and eight years old. Kevin's nephew was less than one year of age when he was diagnosed with cystic fibrosis after presenting with clinical symptoms of the disease about fifteen years ago. There had been other children with CF in the country town that Kevin grew up in so they knew a little about the disease. Anne and Kevin did not choose to have any prenatal testing at all during their first pregnancy and their increased risk of having a child with CF did not occur to them at this point. They went for a genetic counselling session after their second daughter, who had Down's syndrome, was stillborn at 33 weeks. They were offered CF carrier testing and, while Kevin was shown not to carry the familial CF gene mutation, it was revealed that Anne was a CF carrier. During her third pregnancy, Anne opted to have amniocentesis (she "needed all the information upfront this time") before going on to have a healthy baby girl. Their children have not been carrier-tested.

9 KATH:

Kath is a 50-year-old married woman who performs home duties. Her husband is 54 and they are both on disability pensions. Together, they have six healthy children – two sons, aged 32 and 19, and four daughters, aged 30, 28, 17 and 15. The oldest three children have moved out of the family home. Their eldest son, Nick, and his partner had their first child, Steven, almost eleven years ago and he was identified as a carrier of cystic fibrosis through the Guthrie test. Further carrier testing of the family at the time revealed that Steven's CF gene mutation could be traced back through Nick to Kath. Two of Nick's siblings were shown not to be carriers of the familial CF mutation; however, the youngest three siblings were not tested.

10 DAWN:

Dawn is a 33-year-old woman, formerly a dental nurse but now a stay-at-home mum looking after her six-month-old daughter, Mary. She lives with her 32-year-old partner who is a concreter by trade. Three and a half weeks after her birth,

Mary was identified via the neonate screening program as being at high risk for cystic fibrosis. Dawn was advised by telephone that her daughter was either affected with the disease or simply a carrier, and that she was being recalled for a sweat test. Dawn knew little about the disease but was greatly relieved to find out that Mary was only a carrier. Neither Dawn nor her partner had a family history of the disease but genetic testing revealed that Dawn was a carrier. Her partner has not yet been carrier-tested but she thinks he will be eventually. Dawn's father also had testing but he was not shown to be a carrier. Dawn's mother did not worry about being tested because her GP said it was of no real relevance, and Dawn's sister does not see it as necessary for either herself or her children to have this information at this stage.

11 LEANNE & ROB:

Leanne is a 31-year-old teacher on maternity leave, having given birth to her first baby, Amy, seven months ago. Her 35-year-old husband, Rob, is a cartographer who works from home. Leanne had forgotten all about the Guthrie test by the time she got a call from the paediatrician saying Amy had tested positive and could possibly have CF. They had heard of CF but knew little about it. In the end, a sweat test revealed that Amy was only a carrier. Carrier-testing identified Rob as the carrier of the gene that Amy has, while Leanne was not shown to be a carrier. Rob understands there is a high chance that his siblings are also carriers but has not told them of their risk. He believes his brother is unlikely to have children but says he will definitely tell his sister because she plans to have children in the next two years.

12 VICKI & TIM:

Vicki is a 28-year-old registered nurse on maternity leave. Her partner, Tim, is a 30-year-old business development manager, and they have a healthy seven-month-old baby called Zane. They have a friend with CF (she grew up in the same country town as Tim) who doctors said was not going to live beyond nine years of age but she is now married. When Zane was three weeks of age, Vicki received a phone call from their private paediatrician who informed her that Zane had "come up with one of the genes for CF" and needed to come in for further testing. Well over a week

later, Zane was sweat-tested and they were “very excited” to find out that he did not have the disease. After talking to family, they found out that Tim’s cousin was a CF carrier but no one had told them. For this reason, they thought Zane must have got the CF gene mutation from Tim but testing revealed that Vicki carried the CF mutation that Zane has, not Tim. Vicki has two sisters – one chose not to be tested because she does not like needles (but said her own children can look into it if they want to), and the other was tested and shown not to be a carrier.

13 TANYA:

Tanya was formerly a registered nurse but is now a full-time mother and student. She is 27 years old and her husband is 32. They have two healthy children – a daughter, Georgia, who is four years old, and a son who is twenty months old. Georgia was born one month premature and had only just come home from the hospital when the pædiatrician called Tanya to tell her that the Guthrie results were back and her daughter had shown a positive result for a CF gene mutation. Sweat-testing revealed that she was only a carrier of the disease. Her parents both had a blood test while they were at the hospital and her father was shown to carry the CF mutation which had been passed on to Georgia, while Tanya was shown not to carry one of the common CF mutations. Andrew’s family did not have a known family history of CF and his parents did not have a carrier test. His mother, however, did urge her other son to have a genetic test before he was married but he resisted the pressure and said it was not an issue for him at the moment. Georgia’s baby brother was not recalled following the Guthrie test so, while he is healthy, it is not known whether he carries a CF gene mutation like his sister does or not.

Informed consent considerations

The ethical researcher views informed consent as an ongoing process. The nature of in-depth, qualitative interviewing makes it impossible for both the researcher and the participant to fully anticipate all that will emerge in the conversation between them. The participants were advised that they could choose not to discuss an issue or even stop the interview at any time if they preferred. All were assured

that their contribution to the study was highly valued and that their interview data would remain anonymous and confidential. During the interviews themselves, I openly reflected upon what the participants had been saying as it was hoped this reflective process would leave the participants feeling satisfied that their intended meaning was clear. Notwithstanding this, they were informed that if in hindsight (perhaps after viewing their interview transcript) they felt regret that they made particular statements and, for any reason, wished to retract them then they would be removed from the transcript accordingly. Having read the Letter of Introduction (see Appendix 8), all participants were required to sign the Consent Form for Interview (see Appendix 9) prior to its commencement.

The interviews

The interviews were unobtrusively tape-recorded and very brief notes were taken when necessary. My aim was to make the participants feel comfortable and 'at ease', hoping that the interviews would be akin to natural, informal conversation. For this reason, participants were asked to nominate an interview location where they felt most comfortable – for almost all of them this was the lounge room or dining area in their own homes, except one mother who preferred to meet with me in a consulting room at the hospital where her child receives care.

An Outline of Interview Questions (see Appendix 10) was developed in order to keep the conversation focussed on the issues related to the research problem. However, it was not prescriptive as an interpretive 'openness' was essential and there needed to be flexibility so the participants could speak about the issues that were most salient to them. Additional questions were included at the end of the Outline of Interview Questions that covered issues I would delve further into if the opportunity arose in any of the interviews.

Immediately prior to meeting with each participant, I read over the Outline of Interview Questions and some hand-written notes I had made about the in-depth interviewing technique and examples of probing questions to keep in mind:

What was it like?

What were you thinking?

How did you feel about that?

Tell me about...

And then what happened...

What did you tell other people about that?

When I met with the participant(s) on the day of the interview, I introduced myself and showed my student card. I accepted a cup of tea or coffee when it was offered and, having sat down in the area where the interview was to take place, I provided the Letter of Introduction from my supervisors at the time (see Appendix 8) and a copy of the Information Sheet (see Appendix 7) for the participant(s) to read over again if they wished. The tape-recorder was set up while they did this. After the Consent Form for Interview (see Appendix 9) was discussed and signed and any questions or concerns that arose were addressed, the interview was commenced. At this point, I emphasised that the informant(s) should not assume that I know anything at all about what their experience has been like for them, that I would like to hear about it in detail, and that they are the only experts in how they feel and in recalling their own experience.

In-depth interviewing: principles and technique

In qualitative research, the interpersonal skills of the researcher are relied upon as the primary research tool and are crucial to the creation of quality data as the basis of the research project (Minichiello et al., 1995, p.131). However, the researcher should also possess technical knowledge about how best to co-produce rich data with the informants. In order to prepare for the interviewing component of this study, I attended research seminars to learn from the interviewing experiences of other qualitative researchers and I read relevant sections from *In-Depth Interviewing* by Victor Minichiello et al. (1995) and *Researching Lived Experience* by Max van Manen (1990).

In their introduction to in-depth interviewing, Minichiello *et al.* (1995, p.12) state that a primary focus of this kind of interview is “to understand the significance of human experiences as described from the actor’s perspective and interpreted by the researcher”. In order to “[reduce] the possible distorting effect of symbols and language which are not part of [the informant’s] everyday usage”, the authors recommend that “we try to retrieve the informant’s world by understanding their perspective in language that is natural to them” (Minichiello *et al.*, 1995, p.68). For this reason, as well as to build a good rapport with the informant, I always endeavoured to use plain English and refrained from using technical or scientific jargon.

Minichiello *et al.* went on to describe in-depth interviewing as

conversation with a specific purpose—a conversation between researcher and informant focusing on the informant’s perception of self, life and experience, and expressed in his or her own words. It is the means by which the researcher can gain access to, and subsequently understand, the private interpretations of social reality that individuals hold. This is made public in the interview process (1995, p.61).

They emphasise that the in-depth interview is an encounter **between** researcher and informant, where any power differential is minimised (unlike the imbalance of power that exists between the roles in survey methods). It is the informant’s account that is highly valued and the focus is on this rather than on the researcher’s perspective as the valid view (Minichiello *et al.*, 1995, p.68). The researcher does, however, have the ability to construct a view of the social reality of the informant through the process of listening to their verbal account and subjecting it to an interpretive analysis (Minichiello *et al.*, 1995, p.69).

The interviews that I conducted as the basis for this research project are best described as a blend of the semi-structured and the unstructured interview (Minichiello *et al.*, 1995, p.65). In describing the various interview models, Minichiello *et al.* (1995, p.65) state that the unstructured interview is akin to a normal everyday conversation but that it is always a controlled conversation geared

towards the research interests. The level of control exerted by the researcher is minimal but still present to keep the discussion relevant to the research problem. In this study, the mode of asking questions followed the unstructured interview process but an Outline of Interview Questions (without a necessarily fixed ordering of questions) was used to stimulate discussion around particular topics, as would usually be the case in semi-structured interviews (Minichiello et al., 1995, p.65). The wording of the questions was not strictly adhered to; deemed to be of importance were not the questions in themselves but the issues central to the research question that they opened up for the informant to reflect upon and discuss. Questions were omitted when the issues they pointed to had already largely been covered earlier in the interview.

The Outline of Interview Questions contained what are known as 'primary questions', which were used to begin the interview and introduce new topics, while 'probing questions' were impromptu and used as follow-up questions (Minichiello et al., 1995, p.90). Probing questions are important in this kind of interview and were employed to ensure that I did not take for granted the common sense understandings that people share because there was always the chance that I interpreted these differently to the informant (Minichiello et al., 1995, p.89). Probing questions were also asked in order to elicit information more fully when an informant's response was too general, vague or ambiguous. However, I was also informed by van Manen that there would be times when patience and silence were equally effective and, in some instances, more tactful ways of nudging the informant to proceed with, and deepen, the story they were telling (1990, p.68).

The Outline of Interview Questions was necessary in ensuring that largely the same issues were covered in each interview but a recursive model of interviewing¹²⁴ was

¹²⁴ The 'recursive model of interviewing' refers to a form of questioning whereby the process of conversational interaction itself is relied upon, "that is, the relationship between a current remark and the next one" (Minichiello et al., 1995, p.80-81). The researcher chooses how recursive they want the interview process to be, which includes deciding "to what extent prior interaction in an interview session should be allowed to determine what is asked next" and also the extent to which the experiences related by informants in previous interview sessions be allowed to influence the questions asked in later interviews (Minichiello et al., 1995, p.80-81).

used, which allowed for a more conversational model and recognition of the fact that each informant and situation is unique (Minichiello et al., 1995, p.80). At times, I allowed the interviews to go off on seeming tangents to preserve the feeling of a natural flow of conversation but these digressions were often useful and sometimes led into another topic of interest related to the research problem. When discussion on a particular issue seemed to be exhausted, I referred back to the Outline of Interview Questions in order to 'kick-start' the conversation again.

Max van Manen warns that when embarking on the 'conversational interview', the researcher must remain oriented to the fundamental question they are wishing to answer to ensure that the interview process remains disciplined (van Manen, 1990, p.66). In line with his advice, interviews consisting of "lots of short (too short) responses to long-winded or leading questions by the researcher" were avoided, while rich interview data containing "sufficient concreteness in the form of stories, anecdotes, examples of experiences, etc.," were sought to prevent an "[indulgence] in over-interpretations, speculations, or an over-reliance on personal opinions and personal experiences" on my part (van Manen, 1990, p.66-67). I followed his recommendation that "it may be helpful to be very concrete" – asking the informant "to think of a specific instance, situation, person, or event" – when wanting to get to the bottom of what a particular experience is like (van Manen, 1990, p.67). This facilitated the process of focussing on the person's concrete experience and opened up a way for their whole experience to be explored more fully.

In qualitative interviews, I consider it crucial that one becomes acquainted with the 'referential context of significance'¹²⁵ of the informants in relation to the issue under study. The more holistic nature of interpretive inquiry invites that this attention be paid to the context surrounding the experience under study. In this case, the referential context of significance included looking at the impact that

¹²⁵ This is a Heideggerian notion and has been explained thus: "Rather than understanding our world through the laws of science or through some god, individuals make the world intelligible by participating in a social context, a world, which has certain customs embodied by and expressed through [the 'They']" (Lemay & Pitts, 1994, p.51).

cystic fibrosis as a disease has had on each informant's family – this being important for gaining a thorough understanding of the nexus of knowledge, beliefs and attitudes that their carrier testing experience was founded upon. I started each interview by exploring the participant's referential context of significance with regard to cystic fibrosis and the way that it was introduced into their consciousness. In this way, I believe the interview was opened up such that the participant's experience of carrier testing could be grasped in a highly contextualised way. It meant that their CF 'horizon of significance'¹²⁶ had been brought to the foreground such that we could engage with this together as we forged through the terrain of the more specific areas of interest under study; a dialectic was created between the dual circumstance of being-a-carrier (or, sometimes, of not-being-a-carrier) and parenting-a-carrier, as these are situated within the participants' referential context of significance.

Commencing each interview with a question around when the informants had first heard of the disease, cystic fibrosis, created a comfortable opening for the conversation to begin. It launched the interview straight into their family's or their own personal story, or led into background information on preconceived ideas they may have had about CF before it affected them in a more personal way. I used the funnelling method of questioning – starting off with a question of a broad nature and then, as the informants became more engaged in conversation, guided them towards more specific issues with questions that narrowed the focus of the interview (Minichiello et al., 1995, p.84). At this point, more searching questions could then be asked directly about the issues under examination. I chose to utilise this strategy because it puts the informants at ease early on in the interview and allows the interview process to be "more relaxed and non-threatening" (Minichiello et al., 1995, p.84). However, the story-telling method was also used to encourage the informants into 'telling the story' of their experiences. This was done through the way questions were asked but also by encouraging the informants to talk in

¹²⁶ Merleau-Ponty talks about what he means by a 'horizon of significance' in this way: "[A]round our initiatives and around that strictly individual project which is oneself, [we recognize] a zone of generalized existence and of projects already formed, significances which trail between ourselves and things" (1962, p.450).

more detail about how various events occurred. One strategy was to ask for examples of the generalisations that they made while another was to show that I was receptive to listening to their stories by not hurrying their answers, by engaging in preliminary chitchat and also by maintaining and displaying interest in detailed accounts from the outset of the interview (Minichiello et al., 1995, p.86).

A word on computer-assisted 'analysis'

I chose not to use software which purportedly allows computer-assisted 'analysis' of large quantities of qualitative data for several reasons. My main reason for doing so was that it cannot analyse the data at all, rather it can only manage it. Another reason was that it takes considerable time to learn to manage the program and become proficient in its use. A possible outcome of this is that the

emphasis on process rather than content may result in a loss of interaction with the data, which then becomes one step further removed from the researcher (Grbich, 1999, p.251).

I also feel that, in some cases, researchers analysing qualitative data probably choose to use 'analysis' software under the belief that it fosters the appearance that they have taken a more objective approach to their data analysis; I thought it important to resist this notion as both nonsensical and unachievable, and because I did not want to project an aspiration towards objectivity. In later reading, I found that Grbich (1999, p.253) refers to this concept as 'reification'. The term is based on the concern within circles where qualitative computing packages have come under criticism that the computer has become reified as "a system of *self-regulation*: a preferred way of defining the social world¹²⁷ concisely and logically, where individuals become depersonalised and events neutralised" (Grbich, 1999, p.253).

And so, instead, an iterative approach to the entire analytical process was employed – I immersed myself in the data and went over it repeatedly, continually revising themes in light of my increasingly refined appreciation of the power of the existential concepts to explain my data.

¹²⁷ Here, Grbich cites the work of Dupuy, J. and Dupuy, P. (1980).

Analysis and interpretation

Immediately after each interview, I recorded overall impressions of the interview for future reference. All interview data was transcribed *verbatim*¹²⁸ prior to analysis. Data collection and preliminary analysis occurred concurrently as this enabled emerging sub-themes¹²⁹ from earlier interview data to be explored further in subsequent interviews. Having transcribed the first four interviews (Stacy, Henry, Rose, Don & Betty), I reflected on the data as a whole and began to document salient sub-themes that had arisen thus far. My readings of existential phenomenology and some critical and feminist theory shaped the way I viewed the data at that point. In particular, my thoughts were organised around the phenomenological notions of Lived Time, Lived Space, Lived Body and Lived Relation to Other.

I organised the emergent sub-themes under the following broad topic areas:

Themes related to parenting a CF child and CF siblings;

Themes involving our technological and scientific approach to disease – our desire to know, to predict, to control (looking at the way society and medicine shape these parents' expectations and attitudes);

Themes directly involving carrier status;

Themes related to ways of living with CF; and

Themes involving the genetic factor (i.e., that are particular to CF being a genetic illness).

I took a 'selective reading approach'¹³⁰ to the next three interviews (Wendy, Josie, Cindy), identifying additional sub-themes and beginning to sort specific sections of data according to the different sub-themes.

¹²⁸ I transcribed the first seven interviews myself but paid to have the last six transcribed by an assistant.

¹²⁹ I use the term 'sub-theme' to refer to preliminary themes identified in the data. In my mind, these early themes of a kind were not all necessarily linked to specific existential concepts but rather, at that point, were all seen as giving meaning to the experience of parenting a child who is, or could be, a CF carrier.

¹³⁰ In *Researching Lived Experience*, Max van Manen describes the selective reading approach as one of three approaches toward "uncovering" or "isolating" themes (1990, p.92-93). In this particular

The interview with Cindy was a pivotal one. It was in this interview that I was able to pinpoint the basis for the as-yet unformulated concern that I had with the philosophical framework I had been planning to use, phenomenology. It was in the interview with Cindy that the carrier 'state' was most eloquently revealed as one of **knowing**, not **being** (see section in Chapter Five: The Carrier 'State' Is One of Knowing, not Being) – and this largely resonated with what had, and would continue to emerge, in the other interviews. So, having now been immersed in the data, for reasons covered in Chapter Two (see section: The Lure of Phenomenology) I no longer felt that what the informants were telling me about their experience suited a phenomenological investigation. Instead, the data – replete with the dilemmas and paradoxes of human existence – pointed wholeheartedly to existentialist philosophy as the most fitting explanatory device. Since existentialism and phenomenology are closely aligned (in terms of their shared philosophical underpinnings), a revision of the 'Outline of Interview Questions' at this stage of the study was neither necessary nor desirable. I went on to complete the interviews but postponed analysis at this point and began reading existentialist philosophy in order to 'lift out' the existential concepts that could be used to interpret my data.

Having read the literature, written the methodology chapter and explicated an analytical framework based around a repertoire of existential concepts, I needed to re-familiarise myself with the interview data. Armed with a fresh understanding of these concepts, I listened to each interview recording again (in chronological order) and made hand-written notes using the 'detailed reading approach'¹³¹. Having completed each one, I scanned the notes again and jotted down the most telling sub-themes from each interview. I did a summary page for each interview, outlining the sub-themes and making a very brief note against each. This gave me a broad overview of each and allowed me to compare and contrast the interviews more readily. From these sub-themes, I identified the very broad overarching

approach, he said "we listen to or read a text several times and ask, *What statement(s) or phrase(s) seem particularly essential or revealing about the phenomenon or experience being described?*".

¹³¹ In this approach, "we look at every single sentence or sentence cluster and ask, *What does this sentence or sentence cluster reveal about the phenomenon or experience being described?*" (van Manen, 1990, p.93).

themes ('grand themes') and catalogued the sub-themes under these accordingly. Once this was done – and closing off the hermeneutic circle¹³² – I went back into the data to exemplify the grand themes. This involved re-reading each interview and, sentence by sentence, lifting out all data relevant to the list of sub-themes I had created for that interview. At times, analysis of earlier interviews was revised in light of a newly-identified sub-theme in a later interview. All in all, this was a systematic process whereby every sub-theme from every interview was now documented and backed-up by its corresponding data.

Four different concept maps were then created to depict analysis of the interview data from the various 'groups' of participants diagrammatically. I applied my knowledge of existentialist theory in order to cluster sub-themes and group them under a grand theme – the interviews in which these arose were then mapped against them. The 'groups' of participants as differentiated for this purpose were:

CF (the disease) in the nuclear family¹³³

CF (the disease) in the extended family¹³⁴

CF (the disease) 'survivors'¹³⁵

CF (the gene) in the family¹³⁶

The grand themes were mapped to corresponding positions on the different maps to assist with examining commonalities and differences in sub-themes between the groups: all contained some sub-themes which originally sat under the grand themes of THE GENETIC FACTOR, RESPONSIBILITY & FREEDOM, INFORMATION,

¹³² The hermeneutic mode of understanding derives from the science of biblical interpretation but is applied today to all kinds of other texts and unwritten sources, such as "human practices, human events, human situations—in an attempt to 'read' these in ways that bring understanding" (Crotty, 1998, p.87). The 'hermeneutic circle' is a consistent theme in hermeneutic theory but can be conceptualised in different ways. Here, I am describing the kind of understanding that is developed out of what is already understood and then used to return to and "illuminate and enlarge one's starting point" (Crotty, 1998, p.92).

¹³³ The informants are talking about children who have siblings with CF and have grown up in the same household with them.

¹³⁴ The informants are talking about children who have cousins with CF and have grown up in different households.

¹³⁵ The informants have lost (child or teen) siblings to CF and are here talking about their own children who may be carriers.

¹³⁶ The informants are talking about children who were identified as CF carriers via the neonatal screening program; there is no history of the disease itself in these families.

TECHNOLOGY and LIVED TIME. However, upon further analysis, these grand themes were re-worked, becoming EVALUATING GENETIC INFORMATION, GENETIC INFORMATION AND ITS POSSIBILITIES, GENETIC INFORMATION AND ITS POWER 'RELATIONS', LIVED TIME AND TIMING, OUR CONTINGENCY, OUR FACTICITY AND OUR TRANSCENDENCE, AND TECHNOLOGY. Another grand theme, DISEASE RISK, featured prominently in the 'CF (the gene) in the family' group because all the children identified as carriers by the neonate screen had initially been flagged as at-risk for the disease. Of course, the grand theme of LIVING WITH THE DISEASE was covered extensively in all groups of participants except the 'CF (the gene) in the family' group, and LIFE AFTER THE DISEASE, by its very nature, could only be present in the 'CF 'survivors'' group. While conversations around LIVING WITH THE DISEASE and LIFE AFTER THE DISEASE were an important way for me to engage with the informants and orient myself to the informants' 'referential context of significance' around CF, most of this data itself falls outside the scope of this thesis and will be analysed and published separately. A grand theme around MEDICAL MATTERS also emerged in all groups and was closely tied to LIVING WITH THE DISEASE and TECHNOLOGY. But, for neatness, the few sub-themes from MEDICAL MATTERS that will be presented here – those related to the genetic aspect of cystic fibrosis – were later slotted under other grand themes where they 'fit' equally well.

'Displaying' my interpretation of the data

In this study, I relied on the voices of parents from all walks of life. And, by using the pronoun 'our' instead of 'their' when talking about the experiences of the participants, I chose to embed myself in the group under study instead of distancing myself from them. This was a conscious epistemological choice (Collins, 1997, p.199).

Only the context of each piece of data displayed can provide us with its probable meaning for the relevant actors (Duster, 1990, p.137). For this reason, I created an overall précis of each participant in the form of a 'biography' for ease of reference by the reader (see section: Biographies of Participants). In some interviews, an overall existential 'tune' was clear (one theme dominated or stood out vividly from

the rest of the text) and, in these cases, specially selected sections of data that best illustrated and encapsulated the entire interview were presented alongside other data in the same thematic area (i.e., Josie's Tale, Cindy's Insight). The "sententious phrase" that best captured "the fundamental meaning or main significance of the text as a whole", at least with respect to the dominant theme, was chosen as the title for that theme¹³⁷.

I found that the sub-themes and grand themes that lent themselves to an existential analysis generally were spread consistently across the different groups of participants, reflecting commonality in their experience. Because most sub-themes were not peculiar to a particular group of informants, the structure of the analysis chapters was not organised according to these groups but rather around the grand themes that tended to pervade all of the interviews. All data chosen for inclusion in the 'display' here (i.e., to be presented in the analysis chapters in this thesis) were linked to an existential concept.

The data was discussed in terms of its relation to the existential concepts discussed in Chapter Three. Where linked sub-themes contained a set of data that resonated with an existential concept, these were merged to form a theme that would be 'displayed' in the Analysis within the relevant grand theme (e.g., Stacy's guilt and feelings of being defective and Kath's consciousness of being stigmatised both spoke of a shame that was redolent of *The Look* and that is why these were discussed together – see section: Shame and Stigma Associated With Having 'Something Wrong'). The most telling pieces of data that would best exemplify each theme were included, as well as data that was contradictory or nuanced.

¹³⁷ I borrow from van Manen here, who says that "[i]n the wholistic reading approach we attend to the text as a whole and ask, *What sententious phrase may capture the fundamental meaning or main significance of the text as a whole?*" (1990, p.93). While he suggests we then "try to express that meaning by formulating such a phrase" (1990, p.93), I chose to lift a telling phrase from the data itself.

As I wrote up the Analysis, I created another concept map to show how the themes were arranged within the chapter, once written¹³⁸. I then had a pictorial overview of how my analysis 'looked' and this continued to assist with finalisation of the analysis chapters, the argument I was building and, hence, the overall thesis. And so, an overall story was told through a collection of grand themes by now composed of themes that were exemplified by data given significance by an existential concept. And while the existential concepts explained the data, the data itself had now become illustrative of the concepts.

Being 'straight up'¹³⁹ about the methods used

All research methods used in the quest to understand people have some shortcomings, and in-depth interviewing is no exception. However, rather than frame these shortcomings as 'limitations' – which would seem to imply another approach could 'do better' – it is preferable to acknowledge upfront what cannot be achieved using this method and affirm that I believe I proceeded in the best way possible given the question I was trying to answer.

When using the interview method alone, the researcher is only able to gain a verbal account of what the informants think and do so there is really no check on whether what they say is self-deception or even "interviewer-deception" (for socially desirable responses cannot be ruled out) (Minichiello et al., 1995, p.33). What the possibility of *Bad Faith* reveals is that "no single perspective of self-reflection can pretend to yield a complete story" (Cooper, 1990, p.118). At best, the data is solely self-reflection and hence I cannot hold that it has the potential to yield the 'complete story' of people's experiences.

It also must be acknowledged that another researcher investigating this same research question, interviewing the same participants and using the same analytical framework would have come up with a varied interpretation of the data from mine.

¹³⁸ Viewing the updated Table of Contents was another way of monitoring how the themes were juxtaposed in relation to each other at this point.

¹³⁹ A colloquial term used to denote someone who does not skirt about issues but is, instead, upfront, direct and honest.

Without reservation, I state that **this thesis contains my own interpretation of the data that I co-produced with the participants in the study.** The provenance of an interview transcript, while capturing the thoughts of the informant, is not theirs alone – once existing as a text, it is equally open to interpretation by another¹⁴⁰. On this, I refer to Sartre who invoked Husserl in noting

that my consciousness appears originally to the Other as an absence. It is the object always present as the *meaning* of all my attitudes and all my conduct—and always absent, for it gives itself to the intuition of another as a perpetual question—still better, as a perpetual freedom. [...] My reactions, to the extent that I project myself toward the Other, are no longer for myself but are rather mere *presentations*; they await being constituted as graceful or uncouth, sincere or insincere, *etc.*, by an apprehension which is always beyond my efforts to provoke, an apprehension which will be provoked by my efforts only if of itself it lends them force (Sartre, 1958, p.61-62).

In reading this passage, we learn that we do not have ultimate control over how the Other interprets our words and our actions. We may make certain representations as to what the meanings of these are but it lies with the Other to interpret what we say and do as they see it. The weight the Other gives to the ‘face value’ of our words and actions will depend on how sincerely they view our ‘efforts’ (i.e., how much ‘force’ they lend our asserted meaning). I was the Other for my participants. Without doubt, I was constantly required to make ‘micro judgements’ as to the sincerity and the meaning of their words and stated experiences throughout the entire interview and analytic process. The job of interpreting their words as captured in a text lay wholly with me.

According to Grbich (1999, p.218), when researchers actively interact with a text in an interpretive manner, they “[place] a frame around a set of data and [lift] it out”.

¹⁴⁰ In fact, Crotty has said that “[i]ncluded in much hermeneutic theory is the prospect of gaining an understanding of the text that is deeper or goes further than the author’s own understanding. [...] Interpreters may end up with an explicit awareness of meanings, and especially assumptions, that the authors themselves would have been unable to articulate” (1998, p.91).

She compares the concept of “framing”¹⁴¹ to Agatha Christie’s fictional character, Miss Marple, who

spends considerable detecting time comparing the situations to which she is exposed with her knowledge of previous events and personalities from her home town of St Mary Mead (Grbich, 1999, p.218).

As the researcher in this study, I had access to my own previous life experiences, as well as to the findings of other researchers who have looked at the psychosocial impact of genetic testing on adults (some of these were discussed briefly in Chapter One – see sections: What Makes Genetic Testing So Different To Other Forms of Medical Testing? **and** The Problem of Knowing: Population Screening for Genetic Carrier Status) and the experiences of clinicians who have written about the ethical issues surrounding genetic testing and, in particular, the carrier testing of children (see sections in Chapter One: Ethical Issues Facing Genetic Counsellors: Dealing With Past, Present and Future **and** Genetic Testing of Children). However, the dominant source of framing came from the philosophical position that I chose to use to inform this research study – the use of an existential framework, from which I distilled and defined concepts that could be used as various interpretive lenses (see section in Chapter Three: The Existential Concepts). Of course, in choosing any one particular frame, other interpretive possibilities are inevitably prevented from emerging; this must be accepted as inevitable in any study that is theory-driven.

It is difficult to articulate the parameters of interpretivist research but one attempt describes the proponents of this research as “shar[ing] the goal of understanding the complex world of lived experience from the point of view of those who live it” (Schwandt, 1998, p.221). It goes on to say that they believe that the world of lived reality and situation-specific meaning is constructed by social actors and that in order to understand this world of meaning one must interpret it. To do this,

¹⁴¹ Grbich (1999, p.218) acknowledges Erving Goffman’s frame analysis as the origin of the concept of “framing”. Goffman (1974) describes frames as “the principles of organisation which govern social events and the actor’s subjective involvement in them” (cited in Grbich, 1999, p.218).

[t]he inquirer must elucidate the process of meaning construction and clarify what and how meanings are embodied in the language and actions of social actors. **To prepare an interpretation is itself to construct a reading of these meanings; it is to offer the inquirer's construction of the constructions of the actors one studies.** [emphasis added] (Schwandt, 1998, p.222)

Accordingly, those who undertake research embedded in the interpretive paradigm acknowledge that they will always leave their own indelible imprint on the data generated through their research activities and the subsequent interpretation of that data. They readily acquiesce that the end-product of their study is in part influenced by **who they are** and **what they know** prior to even commencing the project.

Max van Manen has emphasised this point well. While van Manen's 'how to' guide to conducting human science research focuses on phenomenological research, in parts his comments apply equally well to other kinds of interpretive inquiry and the following excerpt is no exception:

[P]henomenological research does not start or proceed in a disembodied fashion. It is always a project of someone: a real person, who, in the context of particular individual, social, and historical life circumstances, sets out to make sense of a certain aspect of human existence. But while this recognition does not negate the plausibility of the insights gained from a specific piece of phenomenological work, it does reveal the scope and nature of the phenomenological project itself. A phenomenological description is always *one* interpretation, and no single interpretation of human experience will ever exhaust the possibility of yet another complementary, or even potentially *richer* or *deeper* description (van Manen, 1990, p.31).

The upshot of all this, of course, is that I do not claim this study to be the 'be-all and end-all' when it comes to understanding the experience of parenting a child who is, or could be, a carrier of a mutation in the CF gene.

Existentialism rejects scientific determinism so it is not anticipated that the insights an existential approach allows here into an aspect of human existence will be satisfactory to the close-minded positivist. The positivist program

[gives] support to determinism at the expense of human free will, and could be expected to view non-experimental explanations of human behaviour as wilfully obscurantist (Minichiello et al., 1995, p.32).

Further, because it sees human behaviour as governed by universal laws, it does not accommodate the notion that people are free to take some responsibility for themselves (Minichiello et al., 1995, p.32) – the very antithesis of Sartre’s “man is responsible for his passion” (Sartre, 2004, p.350).

Marcel highlighted the contrast between the “philosophical attitude” – “one of ‘admiration’ or wonder before the mystery of existence” – and the “kind of thinking, typical of science, which seeks to grasp and manipulate and use reality, to impose its categories upon reality in an effort to possess it” (Charlesworth, 1975, p.24). The aim of this research project was not to explain or systematise or prove anything (as is the usual concern of a scientist) but instead to give a plausible account of what happens in carrier families – to evoke, to show, to reveal something of what their experience is and something of what that means for health professionals and society (as is the wont of a philosopher) (1975, p.9). The findings from this study will not allow for the prediction or control of behaviour – in fact, an outcome such as this would not be compatible with the philosophical tenets and the values that underpin the entire thesis.

Existentialists apply to life itself Aristotle’s warning that “it is a mistake to seek a greater degree of clarity than the subject matter allows. You don’t look for mathematical precision in moral matters” (Flynn, 2006, p.66). This was remembered here. And because *Ambiguity* is prized by existentialists, I was not compelled to seek consistency amongst the voices in the study and not all tensions in the interview data were necessarily resolved. However, in keeping with the spirit

of the existentialist philosophers, my aim was to strive “to make sense insofar as sense can be made in and out of our contingent world” (Flynn, 2006, p.37).

THE EXISTENTIALIST ‘ETHIC’: a position from which to view the ‘greys’ of life

[T]o say that we invent values means neither more nor less than this; that there is no sense in life à priori. Life is nothing until it is lived; but it is yours to make sense of, and the value of it is nothing else but the sense that you choose. (Sartre, 1975, p.367)

This study has been undertaken with the purpose of illuminating the ethical debate surrounding genetic testing, specifically, the genetic testing of children. It is important to highlight the ‘ethic’ of existentialist thought and show how this can be used as a guide to the way we view dilemmas involving carrier testing and the way we judge the worthiness of decisions as against their adherence to the values of existentialist thought.

An ethic of authenticity

There is a misconception that Existentialism is a “nihilistic philosophy of despair” (Olson, 1961, p.3). In fact, open-minded and avid readers of the philosophy will realise that the existentialists “mastered the technique of reaffirming the value of life while boldly depicting its horrors” (Olson, 1961, p.2). Simone de Beauvoir put it this way: that those

who feel the joy of existence and assume its gratuity (that is, those who joyfully embrace their contingency) will weather the nihilistic storm brought on by Nietzsche’s ‘death of God’ (Flynn, 2006, p.79).

Ethical considerations **do matter** to the existentialists. And it is Sartre’s promise in *Being and Nothingness* (1958) that an ethic of authenticity – with freedom as an

absolute end – is possible. Flynn has explained the existentialists' common take on how an ethic of authenticity relates to us as individuals:

Though each existentialist understands the ethical, as with 'freedom', in his or her own way, the underlying concern is to invite us to examine the authenticity of our personal lives and of our society (2006, p.8).

Flynn said that, for Sartre, this was about "each of us [acknowledging] what we are doing with our lives right now". He went on to say that

we are challenged to own up to our self-defining choices; to make them our own and consequently to become selves by acknowledging what we are. This is a form of Nietzsche's prescription to 'become what you are'. It's a matter of living the truth about ourselves, about our condition as human beings. The inauthentic person, in Sartre's view, is living a lie.

And what is that truth about our condition, and how are we to live it? [...] [T]he truth which the authentic person lives is primarily a way of life, a manner of existing. (Flynn, 2006, p.64)

A philosophy of life: how should I act now?

What must be done, practically? Which action is good? Which is bad? To ask such a question is [...] to fall into a naïve abstraction. (de Beauvoir, 2004, p.417)

The focus of existentialism is on the proper way to act – a **philosophy to live by** – rather than on "a universal set of principles that ought to govern behavior, principles that are formulated and grasped by the rational mind" (Diprose, 2005, p.238) and grounded in the abstract. The interest of the existentialists is held by a more personal notion of 'truth' – a truth of a moral nature. Kierkegaard once said: "the thing is to find a truth which is true *for me*, to find the idea for which I can live and die" (n.d. cited in Flynn, 2006, p.3). For Kierkegaard, it is a matter of appropriating the truth, of making it one's own; his "emphasis is on the 'how' and not on the 'what' of our belief" (Flynn, 2006, p.10), as with the existentialists

generally who focus on the **way** to make ethical decisions rather than debate about **what** the 'right' decision to make in any given situation is.

The existentialists were all, in their own way, "concerned with the 'moral fact'", stressing that "we are awash in obligations and values that are not the logical conclusion of any series of impersonal facts about the world" (Flynn, 2006, p.63). Kierkegaard (2004a) elevated the individual above the universal, asserting that the standard moral rules set by society are not absolute in the sense that they must be followed by all, always. Kierkegaard reified the "purely private endeavour" that is the living out of a "purely personal virtue" based on faith (2004a, p.14). Sartre rejected a rule-based ethics, saying a morality according to these kinds of rules was no morality but only *Bad Faith*. Arising out of Sartrean and Nietzschean existentialism (and possibly inspired by Kierkegaard) was what became known as a "situation ethics", defined as "an approach to moral decision-making that considers each ethical case to be unique and incomparable, except in a general rule-of-thumb manner" (Flynn, 2006, p.35). This approach recognises that it is simplistic to suppose that abstract values may be adequately and uniformly applied to the complex, multi-faceted scenarios that we encounter daily in 'real life'.

Flynn crystallises what making the "right choice" means for the existentialist philosopher:

If one translates a secularized existential truth into the language of the meaning of life, it would imply that there is no 'objectively' correct path to choose. Rather, for the existentialist, after getting clear on the options and the likely outcomes, one *makes* it the right choice by one's follow-through. For the existentialist, such truth is more a matter of decision than of discovery [...] where the decisive move is not purely intellectual but a matter of will and feeling. (Flynn, 2006, p.10)

Sartre compared ethics with art in order to articulate that which is common to the way we may judge the worthiness of a particular moral choice or a particular painting, neither of which is subject to strict rules. Again, we see that it is only **after the fact** that the choice can be judged:

It is clearly understood that there are no *a priori* aesthetic values, but that there are values which appear subsequently in the coherence of the painting, in the correspondence between what the artist intended and the result. Nobody can tell what the painting of tomorrow will be like. Painting can be judged only after it has once been made. What connection does that have with ethics? We are in the same creative situation. [...]

[...] What art and ethics have in common is that we have creation and invention in both cases. We can not (*sic*) decide *a priori* what there is to be done. [...] Man makes himself. He isn't ready made at the start. In choosing his ethics, he makes himself, and force of circumstances is such that he can not (*sic*) abstain from choosing one. (Sartre, 2004, p.361-362)

What Sartre is saying is that we are obliged to devise our law ourselves (2004, p.362); moral values are "the result of our creative 'choices'" (Flynn, 2006, p.43). And it is because of this freedom/ability to create values that Nietzsche promulgated the possibility of a "higher morality", believing an "ethics of freedom" is available to those "free spirits" who have the courage to live according to "life-affirming" moral values (Flynn, 2006, p.40-41).

I refer now briefly to Søren Holm's summary of Løgstrup's¹⁴² theoretical framework for ethics. It said that

[t]he first question to be answered in ethics is not how we can evaluate moral agents or acts from a third personal, impartial perspective, or even how we should evaluate our own acts after we have performed them (Holm, 2001, p.28).

Rather, for Løgstrup, **ethics is "fundamentally about thinking in first person terms" and its first and most important question is "how I, the concrete person in the concrete situation should act now"** [emphasis added] (Holm, 2001, p.28). I contend that most of the existentialists would share Løgstrup's view and it is seen as important here in the analysis of the data in this study because I am examining first person accounts given by 'concrete people' in 'concrete situations'. How did

¹⁴² K. E. Løgstrup (1905-1981) was a Danish theologian and philosopher, mainly inspired by the German phenomenological philosophers. He studied for one year with the physician and philosopher, Hans Lipps, whose work greatly shaped his thought (Holm, 2001, p.27).

they act in relation to others? What ethic were they informed by? And can their accounts/actions, in turn, inform an ethic for genetic service providers?

The willing of Freedom – both for oneself and all others – as the foundation of all values

For to be free is not to merely cast off one's chains, but to live in a way that respects and enhances the freedom of others.¹⁴³

While Nietzsche “emphasized the importance of style over substance”, it was Sartre’s assertion that there is “a ‘universal’ character to moral judgements” (Flynn, 2006, p.78/79). Sartre insists that

the ultimate value, the goal of our endeavours, should be the fostering of the freedom of the individual, by which he means the enhancement of his or her concrete possibilities of choice. [...] When Sartre insists that one must ‘choose, that is invent’, he doesn’t mean simply ‘improvise’. Rather, he is referring to the responsible decision to opt for or against freedom itself. [emphasis added] (Flynn, 2006, p.47)

And this includes willing the freedom of others as well as one’s own:

We want freedom for freedom’s sake and in every particular circumstance. And in wanting freedom we discover that it depends entirely on the freedom of others, and that the freedom of others depends on ours. [...] I am obliged to want others to have freedom at the same time that I want my own freedom. I can take freedom as my goal only if I take that of others as a goal as well. (Sartre, 2004, p.363)

Simone de Beauvoir agreed with Sartre, arguing that “moral worth resides in the way that we relate ourselves to our own freedom and to the freedom of others” (Marino, 2004, p.412). She also believed that “[one’s] freedom is enhanced, not diminished, when [working] to expand the freedom of others”; in fact, that the

¹⁴³ Quote attributed to Nelson Mandela. Printed on a greetings card, c. 2008 by Oxfam Australia Trading Pty Ltd, Kilkenny (SA) © Freedom and Justice. An internet search for the original source for this quote was unsuccessful.

concrete freedom of the individual relies upon their pursuit of what she calls “the ‘open future’ of others” – that is, “the maximisation of their possibilities” – as well as that of their own (Flynn, 2006, p.79/80).

De Beauvoir acknowledged the existentialist ethic as an individualistic one in the sense that

it accords to the individual an absolute value and that it recognizes in him alone the power of laying the foundations of his own existence. [...] [T]he individual is defined only by his relationship to the world and to other individuals; he exists only by transcending himself, and his freedom can be achieved only through the freedom of others. He justifies his existence by a movement which, like freedom, springs from his heart but which leads outside of him.

This individualism does not lead to the anarchy of personal whim. Man is free; but he finds his law in his very freedom. First, he must assume his freedom and not flee it; he assumes it by a constructive movement: one does not exist without doing something; and also by a negative movement which rejects oppression for oneself and others. (de Beauvoir, 2004, p.433)

In short, to follow the existentialist ethic is to opt for *Freedom* for oneself and for all others ... and this is being *Responsible*.

One could not be blamed for being puzzled that what is, on the one hand, an individualistic ethic still, on the other hand, places an emphasis on the responsibility that the individual has towards others. I believe that Løgstrup’s analysis of the phenomenology of ‘the entanglement’ highlights the inescapability of the common stake that we share with others in the attainment of freedom. Holm summarises this analysis:

According to Løgstrup it is a basic ontological fact about human existence that we are always already entangled or intertwined with, and in the life of, other human beings. This entanglement is not a contingent attribute of human existence, something that happens to us, but that could just as well not happen. The entanglement of our lives is there ontologically prior to our

constitution as individual persons. [...] [H]uman individuals only come into being already entangled in the lives of others. To exist as a human being is always to exist with others. (2001, p.27-28)

The stated basis of Løgstrup's ethics is what he believes is the consequence of this Being-with nature of our existence:

Because we are entangled I always hold some part of the other person's life in my hand. Some part of the other person's life is dependent on how I act in this situation. The mere fact of entanglement thus creates an ethical demand on me to act in a way which takes care of that part of the life of the other that I hold in my hand. (Holm, 2001, p.28)

It is Løgstrup's belief that our entangled situation creates an ethical demand that is "unilateral and radical"; that "[i]t is not a question of reciprocity" (Holm, 2001, p.28).

The unilateral dimension of this moral imperative is not compatible with what de Beauvoir says – for if one's freedom is enhanced through working towards maximising the possibilities of others, then reciprocity is sufficiently established. We protect our own freedom by protecting that of others. This is self-serving. It is ironic that the very nature of Løgstrup's 'entanglement' itself would seem to back this: one should not forget that the entanglement means some part of our **own** life is also held in the hand of the other – any "ethical demand on me to act in a way which takes care of that part of the life of the other that I hold in my hand" would, one presumes, apply equally to the other in their conduct towards me. For as long as I am part of a community, 'taking care' of the part of another's life that I am involved with can never stop at being a unilateral gesture – even if that is my intention in the moment of 'caring'.

Furthermore, there are different views on the evolution of morality. The typically existentialist view of morality was highly influenced by Nietzsche's *On the Genealogy of Morals* (2004) (see section in Chapter Three: *Freedom and Responsibility*). In line with my summary of Nietzsche's discussion of the origins of *Responsibility*, I do not agree with Løgstrup's inference that "the mere fact of

entanglement” creates an ethical demand on us as such. **It is not so much that we should take care of the other (by ensuring we do not constrain their possibilities) but that it serves our own interests (our own Freedom) to do so. Also, as a citizen we are obliged to do so.** Our indebtedness to our community for the protections it offers us and also a fear of punishment as retribution for any wrongs committed by us is the true origin of our *Responsibility* for others, says Nietzsche (2004, p.160/151), rather than it deriving from some kind of moral imperative that naturally arises out of the Being-with nature of our existence.

In summary, there can be no doubt that Existentialism is an individualistic philosophy but it is not unconcerned with social solidarity. It recognises that the life of the individual is ‘entangled’ with the lives of others and that these others necessarily play a part in the likelihood that the individual will be free to reach the heights of authenticity. It also recognises that, due to this entanglement, the individual has the potential to impact on the pursuit of freedom by others. It is for this reason that Flynn labels the philosophy a ‘chastened individualism’, and elaborates by concluding that “the existentialists try to conceive of social solidarity in a manner that will enhance rather than compromise individual freedom and responsibility, which remain non-negotiable” (Flynn, 2006, preface).

Does the existentialist ethic allow us to pass judgement in relation to the choices of others?

In examining the potential of the existentialist ethic to guide the way we behold the dilemmas present in the ‘genetic testing of children’ arena, we must ask whether it provides a lens which allows us to pass judgement in relation to the choices of others. According to Sartre, the answer is ‘yes’ – but only in two ways. The first is that we can make a **logical judgement** as to whether the choice was based on error or on truth:

[I]n certain cases choice is founded upon an error, and in others upon the truth. One can judge a man by saying that he deceives himself. Since we have defined the situation of man as one of free choice, without excuse and without help, any man who takes

refuge behind the excuse of his passions, or by inventing some deterministic doctrine, is a self-deceiver. [...] [I]t is not for me to judge him morally, but I define his self-deception as an error. (Sartre, 1975, p.365)

In other words, we can reprove the choice that is made from a position of *Bad Faith*.

Secondly, we can make a **moral judgement** as to whether the choice honoured a commitment to *Freedom* (the *Freedom* of the individual and the *Freedom* of others):

I can bring moral judgement to bear. When I declare that freedom in every concrete circumstance can have no other aim than to want itself, if man has once become aware that in his forlornness he imposes values, he can no longer want but one thing, and that is freedom, as the basis of all values. That doesn't mean that he wants it in the abstract. It means simply that the ultimate meaning of the acts of honest men is the quest for freedom as such. [...]

[I]n the name of this will for freedom, which freedom itself implies, I may pass judgment on those who seek to hide from themselves the complete arbitrariness and the complete freedom of their existence. (Sartre, 2004, p.363)

But, in the concrete situation of making a decision about the genetic testing of a child, what does a choice 'honouring a commitment to freedom' look like?

The existentialist ethic offers to 'clear' the 'genetic testing of children' minefield

Discussed in Chapter One (see section: Ethical Issues Facing Genetic Counsellors: Dealing With Past, Present and Future) were the elements of a 'principle-based ethics' that are commonly associated with the 'genetic testing of children' debate in the literature, namely autonomy, non-directiveness, informed consent, rights of the parent and rights of the child. Of course, ethical norms from medicine such as beneficence, non-maleficence and justice are always at the centre of the mix too ("Australasian Society of Genetic Counsellors (ASGC) Code of Ethics," 2008). In her book, *Genetic Dilemmas: Reproductive Technology, Parental Choices, and Children's Futures*, Dena Davis says that the limitations of a principle-based ethics are that "it

offers few guidelines for ordering principles when duties conflict" (2001, p.23)¹⁴⁴. There are times when these principles are pitted against each other and there is no guide as to how any particular one can be elevated over and above the others. This system of ethics leaves genetics service providers with no guide for choosing between conflicting interests, which "makes for obvious difficulties and frustrations" (Davis, 2001, p.23). In particular, Davis claims that beneficence (concern for the child's quality of life) and respect for autonomy (concern for the parents' right to make their own decisions about these matters) will "always remain in some tension" within genetic counselling/testing contexts (2001, p.23).

Speaking on behalf of the existentialists, Sartre offers a direct criticism of the 'usefulness' of a system of ethics that is based on abstract principles, again emphasising that commitment to freedom is the only yardstick against which we can 'measure' the value of a decision:

Kant says that freedom desires both itself and the freedom of others. Granted. But he believes that the formal and the universal are enough to constitute an ethics. We, on the other hand, think that principles which are too abstract run aground in trying to decide action. [...] There is no way of judging. The content is always concrete and thereby unforeseeable; there is always the element of invention. The one thing that counts is knowing whether the inventing that has been done, has been done in the name of freedom. (Sartre, 2004, p.364)

In applying this view to the genetic counselling/testing context, we see that attempts to delineate an abstract set of principles to guide parents and professionals through the ethical minefield that surrounds the genetic testing of children is not only misguided but also unlikely to assist in working through real-life scenarios and dilemmas. It reveals that the traditional bioethical approach, with its strong medical emphasis, is insufficient. Each concrete ethical dilemma requires an original approach.

¹⁴⁴ Here, Davis cites the work of Jonsen (1994).

Simone de Beauvoir looks at the special ethical situation in which we find ourselves when we are called on to make decisions on behalf of another/others, as is the case when parents and health professionals are charged with looking after the best interests of the child:

[T]he good of an individual or a group of individuals requires that it be taken as an absolute end of our action; but we are not authorized to decide upon this end *à priori*. The fact is that no behavior is ever authorized to begin with [...]. To put it positively, the precept will be to treat the other (to the extent that he is the only one concerned, which is the moment that we are considering at present) as a freedom so that his end may be freedom; in using this conducting-wire one will have to incur the risk, in each case, of inventing an original solution. (de Beauvoir, 2004, p.423)

We hear de Beauvoir advise that in each situation we must act out of what we believe will be for the ultimate 'good' of the other, where she conceives of this 'good' no more narrowly than as 'opening up their concrete possibilities to freedom'. However, she says, we must not presume to decide what that good (for the other) is in advance.

Introducing the existentialist ethic into the 'genetic testing of children' arena offers a novel approach to the examination of ethical dilemmas that emerge there. The existentialist ethic differs markedly from a principle-based ethics and promises to 'clear' the 'genetic testing of children' ethical minefield of the juxtaposed and, at times, conflicting principles that currently comprise it. However, it must be noted that even the commitment to freedom (ultimately) sometimes "[flees] from itself" (freedom in the moment) (de Beauvoir, 2004, p.418). It is for this reason de Beauvoir would say that even Sartre's directive that "[o]ne can choose anything, but only if it is upon the plane of free commitment" (1975, p.367) is ambiguous. She explains that the potential for freedom in the future can justify its denial in the present; that in terms of willing the freedom of others, there are times when it is right to "[reject] benevolence to the extent that the latter thoughtlessly sacrifices the future to the present" (2004, p.418). Such is the "ambiguity of freedom" (2004, p.418), de Beauvoir even sanctions "violence" against another in its name:

It is no more necessary to serve an abstract ethics obstinately than to yield without due consideration to impulses of pity or generosity; **violence is justified only if it opens concrete possibilities to the freedom which I am trying to save**; by practising it I am willy-nilly assuming an engagement in relation to others and to myself [...]. If I find myself in a position to do violence¹⁴⁵ to a child, or to a melancholic, sick, or distraught person the reason is that I also find myself charged with his upbringing, his happiness, and his health: I am a parent, a teacher, a nurse, a doctor, or a friend ... So, by a tacit agreement, by the very fact that I am solicited, the strictness of my decision is accepted or even desired; the more seriously I accept my responsibilities, the more justified it is. That is why **love authorizes severities which are not granted to indifference**. [emphases added] (de Beauvoir, 2004, p.419)

In the context of this study, what “severities”, if any, does the love of the parents for their children authorise in terms of genetic testing-related decisions? According to the existentialist ethic, it would seem depriving their child of an aspect of their freedom and autonomy now could only be justified if the parents believe it will open up other concrete possibilities to freedom for them. What about the genetic counselling professionals? They commit an act of violence by rebuffing the autonomy of the parents but surely this becomes defensible under the existentialist ethic if it is done in the name of safeguarding and maximising the possibilities of the child in question...

Preserving the child's right to an open future

The core theme of Davis' book, *Genetic Dilemmas*, is “how decisions parents make about genetics can expand or limit their children's future possibilities” (2001, p.70). In this way, she sets up a discourse that resonates with that of the existentialists though she does not refer to them explicitly. She too rejects addressing ethical dilemmas via the “conflicting-values approach”, and she attributes this predominantly to her “strong commitments to the primacy of the value of

¹⁴⁵ While she does not say so explicitly, I believe de Beauvoir's use of the word 'violence' here can be taken to refer not only to physical violence but also to the kind of assault involved in temporarily depriving a person of their freedom; their autonomy; their sovereignty over themselves.

autonomy" (2001, p.23). This has strong resonance with the existentialists' call that we actively work to protect and expand the freedom of others. In the genetic counselling/testing context, this must mean maximising the possibilities of the individual. To move the ethical debate around the genetic testing of children beyond the unresolvable tension between beneficence and respect for autonomy, Davis suggests reconceptualising the tension as a conflict between **parental autonomy** and the **child's potential autonomy** – the latter, she says, is what the philosopher Joel Feinberg first articulated as "the child's right to an open future" (Davis, 2001, p.23).

In his discussion of children's rights, Feinberg (1980) points out that there are different classes of rights – some common to adults and children, some that can only be exercised by adults and some that are generally only held by children. His focus is on a particular class of children's rights that he says are held in trust by parents until their children are "more fully formed and capable" (1980, p.125). These "anticipatory autonomy rights" he labelled "rights-in-trust" (1980, p.126/125). All rights-in-trust relate to the child's potential autonomy and, by their very nature, "are to be *saved* for the child until he is an adult" (Feinberg, 1980, p.125). They include virtually all the important rights we associate with adulthood but which must be protected now in order to be exercised later. Feinberg explains that rights-in-trust can be violated and that **the adult who the child is-to-become** is, in fact, "the person whose autonomy must be protected now (in advance)" (1980, p.127):

The violating conduct guarantees *now* that when the child is an autonomous adult, certain key options will already be closed to him. His right while he is still a child is to have these future options kept open until he is a fully formed self-determining adult capable of deciding among them. [...] [These rights-in-trust] are, in effect, autonomy rights in the shape they must assume when held "prematurely" by children. (1980, p.126)

It is all the rights grouped together and categorised as 'rights-in-trust' that constitute what Feinberg calls the child's "right to an open future" (1980, p.126).

Davis agrees with Feinberg that “parents ought not to make decisions about their children that severely and irreversibly restrict their right to an open future” (Davis, 2001, p.27). Feinberg says that the existence of the right of the child to an open future

sets limits to the ways in which parents may raise their own children, and even imposes duties on the state, in its role as *parens patriae*¹⁴⁶, to enforce those limits (1980, p.140). [...] When the state justifies its interference with parental liberty by reference to the eventual autonomy of the protected child, it argues that the mature adult that the child will become, like all free citizens, has a *right of self-determination*, and that that right is violated in advance if certain crucial and irrevocable decisions determining the course of his life are made by anyone else before he has the *capacity of self-determination* himself (1980, p.143).

Feinberg uses legal cases to make his argument but Davis points us to his critique of U.S. court decisions only in order that the underlying ethical basis for his argument is clearly understood; that we see it at work in a concrete context. She emphasises that she is not agitating for new laws that regulate genetic counselling practices but rather that policies and attitudes – for instance “at the level of professional societies and training” – now reflect the ethical stance that she shares with Feinberg (Davis, 2001, p.27-28).

Davis makes no reference to existential philosophy in her treatise but it is clear that the existentialists would approve of the way she conceptualises the issue. If we now re-examine the question posed above about what a choice honouring a commitment to freedom looks like in terms of genetic testing and the best interests of the child (see section: Does the existentialist ethic allow us to pass judgement in

¹⁴⁶ The *parens patriae* doctrine grants power to the state to protect “those who cannot help themselves” (Feinberg, 1980, p.130). Feinberg (1980, p.129) points to a legal interpretation of this doctrine which stipulates that, in doing this, the state must only “decide for [a man] as we assume he would decide for himself if he were of sound mind” (“Civil Restraint, Mental Illness, and the Right to Treatment: Note and Comment,” 1967). Feinberg states that this becomes more problematic when the courts must decide for a child and, as a general rule, “the courts will not be so presumptuous as to speak now in the name of the future adult” (1980, p.129). Notwithstanding this, he says “there are sometimes ways of interferring (*sic*) with parents so as to postpone the making of serious and final commitments until the child grows to maturity and is legally capable of making them himself” (1980, p.129).

relation to the choices of others?), we see that Davis' work has bridged this gap. To honour a commitment to freedom in the context of genetic testing is to avoid restricting the possibilities of the individual unnecessarily; in the delicate context of the genetic testing of children, this means preserving the child's right to an open future above all else.

The existential concepts of *Freedom* and *Authenticity* were explained in Chapter Three (see section: The Existential Concepts). These are at the heart of the existentialist ethic which has been outlined here, and we now have the child's 'right to an open future' as an extension of this. This ethic will be used as a guide for analysing the data produced in this study. It provides a way of looking at what parents are saying and doing in relation to their child's carrier status and gives us a basis against which we can form judgements about their stated attitudes and decisions. We can ask whether the parents in this study are conscious of not wanting to foreclose their children's futures. We can examine whether their actions/attitudes are working to expand or to limit their children's future possibilities.

CHAPTER FIVE: KNOWING GENETIC INFORMATION

INTRODUCTION

Genetic information has **stakeholders** – people who are (or might be) affected by that information. Genetic information also has **gatekeepers** – those made responsible not only for identifying which stakeholders have a right to it, but also for deciding when to tell and how to tell what they know. For this reason – whether they realise it or not – the gatekeepers are in a position of power to influence the way that genetic information is evaluated by the recipient. The choices that health care and genetics professionals, parents and family members make around imparting genetic information to children and other family members have the potential to confine their ‘possibilities’. The *responsible custodian* is aware of this and will ‘care for’ the information in a way that maximises the possibilities of its stakeholders, thereby enhancing their *Freedom*.

The bioethical discourse surrounding genetic testing has tended to focus on issues such as confidentiality, privacy and autonomy; these all reinforcing ideas of secrecy, taboo and individuality – pulling apart families and separating out its members, making them distinct from each other. The family is not viewed as a **unit** but rather as a **group of individuals** who are potentially mistrustful and secretive. While a silence does envelop the shared genetic threat in some families, one often finds that the family as a whole consists of a network of shared knowledge about the illness; its treatment; its severity; the experience of the illness sufferer; its burden on the parents; speculations about the mysterious ailments of ancestors; a shared surprise that this has happened to them; an understanding of its genetic implications for the family as a whole; and a rethink about their relatedness to distant and unknown relatives. An entire extended family is affected by the risk attached to the discovery of a CF gene mutation in one of their relatives but Technology pinpoints which members of the family this affects as individuals. Often, knowledge of the ‘whereabouts of the gene’ in their family has been garnered from various family members who may differ in the extent to which they

are 'in the know'. Family members often go for testing together and are aware of each other's results.

This first analytical chapter looks at what is happening in families when genetic information becomes known.

How is the information being evaluated and incorporated (or not) into a person's sense of who they are? What are adults doing with the information as it applies to them? How is it being used to inform their decisions around pregnancy and prenatal testing? Is it serving to increase their choices or does it sometimes decrease them? How does the power that comes with knowing genetic information exert itself through family relationships? How do the custodians 'care for' the information within the family? What are the expectations of the stakeholders? What motivates the gatekeepers to withhold or release this information? And what informs the manner in which they do this? Is the child's right to an open future being threatened or safeguarded by their parents and health/genetics professionals?

EVALUATING GENETIC INFORMATION

The Gene and Its Relationship to Identity

SHAME AND STIGMA ASSOCIATED WITH HAVING 'SOMETHING WRONG'

The two parents who spoke of being a carrier as having 'something wrong' were also those who felt some level of shame about it; that there was a stigma attached to them.

Stacy wondered whether her daughter may one day feel a little 'defective' knowing that she is a CF carrier because she confesses that this is how she feels about it (21: 46-49). She admitted to negative feelings about her self-image when asked how she felt about the 'carrier' label but she attributed these feelings to the fact that, in her case, being a carrier had resulted in her child being born with the disease:

I thought probably as well I feel a bit guilty being a carrier...I don't know, and then having Bradley and giving it to him. Yeah, but 'defective' is probably the best word... (22: 12-14) [...] If I didn't have a child with cystic fibrosis, it wouldn't worry me...that I was a carrier, and that, you know, I wouldn't feel as defective...as I do because I have a child with CF. (23: 53-55)

It seems she was most conscious of her feelings of being 'defective' when she saw herself through the eyes of other people:

Sometimes when I have to explain [to other people] that two people are carriers [to have a child with CF], it makes me feel like – again, like I'm defective, like there's something wrong with me, I suppose...because I carry it and then Steve carries it as well. Yeah...I don't know – that's probably about it. I just feel defective, a little bit.¹⁴⁷ (23: 20-23)

Her shame before the Other is characteristic of that described by Sartre (1958) in the section on *the Look* in his philosophical treatise, *Being and Nothingness*. Sartre focussed on the example of shame as a mode of consciousness because it is particularly illustrative of *the Look*:

[I]ts structure is intentional; it is a shameful apprehension of something and this something is *me*. I am ashamed of what I *am*. Shame therefore realizes an intimate relation of myself to myself. Through shame I have discovered an aspect of *my* being (Sartre, 1958, p.221).

In Stacy's own account of how she feels about being a carrier, her shame is uncovered in the moments that she is subject to *The Look* and conscious of herself as an object for the Other. It is in *The Look* that she discovers an intimate aspect of her being that she would otherwise not be privy to. Such is the nature of shame as a mode of consciousness that her shame can only be "*shame before somebody*" (Sartre, 1958, p.276), which explains why *The Look* was requisite to her awareness

¹⁴⁷ According to Goffman's way of theorising stigma, Stacy experiences her carrier status as something which makes her a "discreditable" person (1968, p.14). Her 'defect' is not "evident on the spot" so it is "neither known about by [others] nor immediately perceivable by them" (Goffman, 1968, p.14). It is in moments of explaining the cause of her son's cystic fibrosis to others that she feels stigmatised by her carrier status as this is when it becomes 'perceivable'.

of this structure of her being; to knowing herself, her innermost feelings about being-a-carrier. In experiencing her body as objectified, she sees it as 'defective'.

However, she does go on to say that she felt comforted to learn that CF mutations are common in Caucasian populations:

[The doctors] did say as well that it's very common - that one in twenty people are carriers which is probably a good thing to say, that it's not an uncommon gene to carry. [...] [That made me feel a] little bit better, thinking, you know, one in twenty – you think "Shit, there's a lot people out there that are carriers and don't know about it". (23: 45-51)

This reveals that some of her shame about being-a-carrier arose because her difference meant she no longer felt she was 'normal'. However, it eased her mind to know that many others carry this same gene because it implied that many others are similarly 'defective', albeit unwittingly so. It seems that her sense of being 'different' is lessened by knowing that a lot of other people share the same difference. It could even be said that she now sees it as a *normal defective* gene to carry¹⁴⁸ – “not an uncommon gene” – and that this is what makes her feel a “little bit better” about being a carrier. The irony here, of course, is that the more common a disease gene is, the higher the risk of having a baby with the recessive illness since it is more likely a potential partner will also carry that same particular disease gene – a fact not lost on Vicki who had said in her interview:

[Y]ou feel less isolated but then the flip side is: to us, it doesn't seem that uncommon any more. Do you know what I mean? And the chance of [our son] meeting somebody who's also a carrier is a little bit more real (20: 41-43).

¹⁴⁸ Rosalyn Diprose has discussed the social evaluation of identity and difference in the practical applications of genetics (2005, p.247). She points out that the claim “this evaluation is relational (to a social norm), rather than a direct or self-present representation” is consistent with the Hegelian philosophical tradition which says “identity is always *produced* through differential relations”. Diprose continues, “To evaluate differences as defective or inferior relies on the (incorrect) assumption that the standard to which the evaluation refers, the “proper,” stands alone. This assumption also pervades the claim that science can represent difference per se, as if the identity from which the difference differs stands apart from that relation and has an identity in itself” (2005, p.247). We see that what Diprose says here resonates with how Stacy perceives her own 'difference' – her feelings of being either defective or normal are produced through differential relations to others.

Geneticists estimate that everyone carries six to eight recessive disease genes but most of these are so rare that the chances of one's partner carrying the same gene are negligible (E. Haan, personal communication, April 27, 2001).

What has happened here? Let us turn again to Sartre's theorising around *the Look* for the answer:

It is shame or pride which reveals to me the Other's look and myself at the end of that look. It is the shame or pride which makes me *live*, not *know* the situation of being looked at" (1958, p.261).

We are reminded here that the shame Stacy feels in the first instance before the Other is immediate, spontaneous and pre-reflective. But shame does become "accessible to reflection" (Sartre, 1958, p.221), giving her an opportunity to have some level of control over whether she allows herself to submit to this emotion or tries to overcome it. She looks to 'evidence' in the world about her to modulate how she feels – and while the opinions or thoughts of others may influence this, in the end she will be wholly *responsible* for whether she continues to feel shame.

Kath also thought that the word 'carrier' was stigmatising and she too spoke of the carrier state as having 'something wrong':

[I]t does kind of have stigma attached to some people and I'm probably one of them: "Shock horror – I'm a carrier", kind of thing [...] ...something's wrong... [laughs] – which it is. (15: 28-33)

This goes some way toward explaining her reaction when she was informed by letter that she is a carrier:

I remember reading it, hiding it, and not showing it to anyone for a while until I had digested it myself! Isn't that silly? (22: 26-27)

She went on to say that she did not even tell her husband about the results for a couple of days, then she eventually got around to informing her three eldest children:

I had to accept it myself before I could even discuss it and show him and then I think it took a little while to tell [my son and daughter-in-law], and when I was talking to my girls interstate, like probably took me a little while to tell them that, "Hey, you've got to have this", you know, "Blood results are back and I'm the carrier!" [...] I think I thought it was a stigma attached to me because otherwise why would I have hid the letter and I think I hid it and then I kept getting it out and reading it and then hiding it away again, which now sounds stupid but... [...] I mean, hiding the letter wasn't going to change the outcome.¹⁴⁹ (22: 50 – 23: 9)

It seems that hiding the letter and withholding her carrier test result from her family was an attempt by Kath to postpone her sense of shame before them; the Other. Again, Sartre's explication of shame can be used to throw some light on this:

[S]hame [...] is shame of self; it is the recognition of the fact that I am indeed that object which the Other is looking at and judging. I can be ashamed only as my freedom escapes me in order to become a given object. [...] Beyond any knowledge which I can have, I am this self which another knows. (Sartre, 1958, p.261)

For as long as the Other (members of her family) was not "looking at and judging" her, Kath remained free not to recognise herself as the object that she anticipated would be perceived as having "something wrong" and of which she must then feel ashamed.

Kath also said it made her feel different from other people when she first found out about her carrier status but claims she reconciled herself to the information later:

[T]hen you kind of stop and take notice and think, "Oh, you kind of wouldn't have done anything different because you didn't know about it". It is just to know that, um...to know something's wrong with one of your kids, and now an adult before it's found out, it's just a bit of a shock. [...] Especially when you find out you are the one that it came from. (16: 49-57)

¹⁴⁹ What Kath is describing here is typical of Goffman's "stigmatized individual", who may find themselves feeling unsure of how others ("normals") will identify and receive them (Goffman, 1968, p.24).

Like Stacy, once she stopped **living** her shame (pre-reflectively) and began instead to **know** her shame (reflectively), she was able to moderate the impact it had on her own self-image.

In both Stacy's and Kath's shame, we see that people are not totally free to interpret what particular genetic information means to them strictly according to their own set of values; their own ethic. They live in their everydayness in an environment where other people matter, where other people's perceptions and values are an issue for them. Their *Freedom* is tied to the attitudes and opinions of others. Such is Merleau-Ponty's emphasis on this point, he concludes *Phenomenology of Perception* (1962, p.456) by quoting A. de Saint-Exupéry (n.d.): "Man is but a network of relationships, and these alone matter to him."

THE CARRIER 'STATE' IS ONE OF KNOWING, NOT BEING

Two mothers revealed that the knowledge one carries a recessive disease gene need not be a challenge to one's identity at all. To them, it is just a piece of information that will not affect their children unless, and until, they decide to have children.

Cindy's children have not been carrier-tested. When asked how they reacted to confirmation that she carried the same gene that had been responsible for her brother's death, Cindy said there was no discernible reaction from them:

It was no major thing to them because I don't have the disease. But they just know that they could be carriers too, and what that means to them as far as having children. Because, and I'm probably still quite ignorant about the disease, but as far as I know it doesn't mean anything for the rest of your life. You know like, it doesn't impact on any other part of your life if you're a carrier unless you're having children... (7: 18-23)

In terms of the fact that this now means her children have a one-in-two chance of carrying the gene too, she did not express any concerns about how their future partners may view this risk:

'Cos I think you can work through that, like it's not...because it only presents in the person who's got it, for me it's easy to kind of compartmentalise it – like, "okay, I'm a carrier" but all it means is that if I have a child with someone else who's a carrier, there's the possibility of having a child with CF. So it's quite a logical sequence – it's not...there's no sideways stuff...that's just what it is. (12: 10-14)

Besides, we all carry recessive genes for severe/terminal illnesses and, as Cindy said, it is really just a matter of "[who we] hook up with! [...] [she chuckles] How your genes get on really, isn't it?" (12: 52).

In effect, her words tell us that for her there is actually no **being** to 'being-a-carrier' – rather, it is a state of **knowing**. And logic steers her toward keeping the knowledge that she has one identified gene change in perspective:

I would never have looked in my life and thought, "Oh, maybe I feel like this because I'm a carrier of CF". It just has no influence like that. (15: 27-29) [...] So then I think "Well, the rest of our gene...genetic makeup – there must be some reasonable stuff in there that I'm healthy!" D'you know what I mean? [...] [The CF gene] is just part of a big picture, isn't it? (15: 41-46)

Yes – it's only one gene in amongst a mass of genetic material that partly determines who we are, who we *be*!

Tanya is satisfied that being a carrier does not affect her daughter's health in any way (14: 49-53) but she does wonder sometimes how it might impact on her when she has children, even that "she might decide that [it] narrows down who her partner could be" (12: 49-50). And Tanya is glad that she has been informed about her daughter's carrier state despite not seeing it as inevitable that her daughter will, or should, 'use' the information (19: 39-41):

I think like it is good to know – even for knowledge's sake – yeah, 'cos it...I mean, it doesn't obviously have a huge impact on her life at the moment, and may never have. (19: 14-16)

As with Cindy, Tanya interprets what being-a-carrier means for her child as a **potentiality** – a piece of information that may come to bear on her **future** – rather

than a state of **being** that affects who she is **now**. If this is the case, the revelation that a person carries a recessive disease gene need not be a challenge to their identity.

Tanya was adopted as a child and so her comments on the carrier state and its relationship with identity are particularly interesting because the 'genetic testing of children' literature does draw a comparison between "handling genetic information about children and handling information about their adoption" (Clarke, 1998, p.9-10). Tanya contrasted the two of her own accord without prior knowledge of this (12: 47-48). On the relationship between carrier state and identity versus adoption and identity, Tanya gave the following insight:

I think it is a little bit different though in that there's a relationship between you and your parents (adopted or whatever), whereas like [knowing you have a recessive gene] is a bit more abstract. Like, it's sort of something that could happen but probably won't, whereas like being adopted you say, "Well, it has happened" and, you know, you have to deal with it. And so from that point of view, my personal opinion is my parents told me when I was quite young and I grew up with that knowledge and it never bothered me to know that [...]. But I don't think that bringing Georgia up knowing that she's got a recessive gene for CF would really do anything. (13: 31-39) [...] [L]ike I think if you turn say fifteen and your Mum goes, "Oh, guess what? You're not really our biological child", that could like send you sort of reeling whereas like saying, "Well, now that you're old enough to understand, I'm going to tell you that you have this recessive gene and it means blah, blah, blah", it's – to me – it's not the same thing. (13: 55 – 14: 3)

So, whereas Clarke supposes that "[b]oth categories of information are likely to be unwelcome but withholding the information from the child for ever is likely to cause further problems" (1998, p.9), according to Tanya's perspective, revealing carrier knowledge to a person is not life-changing in the way that adoption knowledge can be. It does not have the same potential to change a person's sense of who they are. It does not necessarily present the challenge to identity that perhaps adoption does.

A REVEALED IDENTITY

One mother was 'shocked' and 'shaken' to discover that her adult son is a carrier and that she, in turn, is one too. Far from being a mere piece of information, she very much sees it as a newly-revealed part of their identity – one that has always been there but remaining hidden until now.

In contrast to Cindy and Tanya, Kath's experience provides an alternate account which reeks of 'challenged identity'. Despite no instance of the disease afflicting the family, even the revelation that her adult son is a carrier was a blow to her:

It was "shock horror" at first [...] [because I had] had six kids and they were all healthy and well, and then to find out your first grandchild [was] being tested and we didn't really know what for [...] I don't know... It just...it just really shook me to know that [my son] was a carrier and I didn't know, I suppose – that's probably what it boils down to. (5: 38-48)

Importantly, Kath is not just upset by the discovery that the CF gene mutation is in her family but that her son **was** a carrier and she **did not know**. She is 'shaken' by the fact that this was there all along; hidden; unbeknownst to her. In this way, we see that she frames her son's carrier state as a revealed aspect of his identity rather than a spoiling of it. This resonates with the findings of a study that looked at the processes employed within genetic counselling consultations (Armstrong et al., 1998). The authors concluded that these processes provide the client with a genetic identity but that the way in which they do so constitutes "a reversal of the stigmatising process" – that is,

[i]nstead of a spoiled identity replacing an actual one, the genetic consultation involves revealing an actual identity in the place of a previously presumed one: in effect, the patient's identity is not spoiled but made visible (p.1657).

They go on to say that

[i]t is this disclosure of a "revealed" identity that separates out the effects of genetic disease from other "chronic" illnesses. [...] In genetic disease there may still be stigma, both felt (in terms of

feeling “spoiled” and enacted (for example, from insurance companies), but it has a different basis. Genetic disease differs in as much as it promises to reveal who the individual always has been, not a new addition but a revelation about an underlying identity that had been concealed. (p.1657-1658)

Given this, Armstrong *et al.* state that “genetic counselling reconstructs identity in the past as well as in the future” (1998, p.1653), and Kath’s account confirms that this can also be the case with the carrier state (in the absence of illness).

Kath went on to speak about her experience of finding out she had a brain tumour and then compared this to her experience of finding out she is a CF carrier:

[W]hen I had my brain tumour, you’re still the same person but when you don’t know, you’re fine, and then when you find out you’ve got it, you say, “Doctor, well I want it out” and he says, “Well, it’s been there for that many years, it’s not going to do any more damage” but it’s just like, “Shock horror - you didn’t know before”. And once you know, you do kind of want to investigate or know more (16: 3-8) [...] [B]efore you know [about being a carrier], everything seems rosy and once you know, there is like a big shock to your system – that you want to find out more, you want to know more to know if you have given this to your kids. (16: 19-21)

While Kath’s experience of shock and horror at finding out she is a carrier is quite a departure from the reported experience of others in this study, it is possible that her previous experience with cancer makes her a special case in the context of this study. As with her brain cancer, the discovery of her carrier status is further reinforcement for her that the body can ‘betray’; that she is beholden to the whims of biology and the arbitrariness of nature and it is in this realisation that she experiences *Sartrean Fear*. We all read and interpret our experiences through those that life has already dealt us and it is likely that Kath’s experience of being-a-carrier is filtered through her prior drastic experience of having had her body betray her.

GENETIC INFORMATION AND ITS POSSIBILITIES

Making Decisions Around Pregnancy and Prenatal Testing

CINDY'S INSIGHT: "INFORMATION IS THE CRUCIAL THING"

Cindy's reflections around access to information while making decisions about pregnancy and prenatal testing, which are based on her own life experience and her hopes for her own children, are summarised here.

The interview with Cindy was a telling exemplar of the paradox surrounding the human hope that information, knowledge and genetic testing technologies will bring to an end all reproductive risks and woes. The *Anguish* that information and knowledge can deliver into human existence is captured beautifully by Dostoevsky in his *The Brothers Karamazov*:

Didst Thou forget that man prefers peace, and even death, to freedom of choice in the knowledge of good and evil? Nothing is more seductive for man than his freedom of conscience, but nothing is a greater cause of suffering (2004, p.241).

However Dostoevsky maintains that, regardless of this, paradoxically, humans are still seduced by the desire to control and therefore continue to seek out choices.

The interview with Cindy proceeded in a fashion that highlighted her belief that access to information is key to autonomy in decision-making and fundamental to a person's ability to make choices from a position of empowerment. But then at the end of the interview in an eleventh hour twist reminiscent of the rhetorical style of Sartre's novel *Nausea* (1963) – where one felt convinced by the life-is-utterly-meaningless sentiments of the hero until a late-in-the-piece revelation turned all on its heel and rescued a way for us as individuals to find meaning within a greater meaninglessness – Cindy unwittingly quashes the notion that information is the cure-all when she reveals that she is in fact glad it was not available to her when she herself was pregnant.

According to Cindy, her CF carrier risk was never an issue for her partner (13: 33-38) – it was only a passing thought when she was pregnant with her first baby:

[I]t wasn't something that I thought: "Hang on a minute here. I'm a carrier of CF. What's this going to mean? How are we going to find out about it? What are the possibilities?" Didn't do any of that. (13: 42-44)

She was always aware of the possibility she could be a carrier but had her children in the mid-eighties prior to testing being available. And while she describes herself as having been "pretty stupid and fatalistic anyway" at that point (4: 43) – "I just went ahead [with it]" (4: 42-43) – her account reveals that her thinking at the time was based on a crude observance of logic and probability:

I figured, in my husband's family there had been no CF – but then there hadn't in my mother's and father's either that they were aware of – and I figured I'd be pretty unlucky [to have a child with cystic fibrosis]. (4: 43-45)

In Cindy's words, being "stupid and fatalistic" meant that she went into motherhood without giving it a lot of thought; it was "just something that I believed I was always going to do, not something that I'd perhaps always wanted to do – in the sense of 'had thought about it'" (14: 26-28). In her own opinion, she had not taken seriously enough her heightened risk of having a baby with CF and she accounts for this as having been "too young" at the time (14: 28-30).

After years of understanding that there was a good chance she was a CF carrier but there being no test available, she said it was "good" finally to have a definitive carrier test because she could then say to her children:

"Look, I'm a carrier and you could be a carrier too. And this is what it means. And you can find out if you are." And I haven't taken it much beyond that. Neither of them have...(well, [my son] is nearly seventeen)...but they're not at that stage where they're having a family. But they definitely know that there's a possibility – quite a likelihood that they could be carriers of the disease, and that I am, and that there needs to be another carrier for them to have...they know about recessive genes, how it will present. (5: 35-41)

Her pre-occupation with her children having the ability to access information so that they can make informed decisions is also evident:

And I would be strongly advising them... [pause] I mean, if they came to me and said, "I want to know more about it" or "I want to go and have the test done", I'd say, "Sure. Go and have it done" and definitely if they were thinking of having a family, I would say. And then if they were carriers, I would really like information to be available to them so they would know what it's like to have a CF child; what that means, I mean to know what that feels like, to know what that means as far as time, life span, all of that...so that they could make really informed choices rather than just do what I did which was just go along, go along and go along [nervous laugh] and I was lucky – really, it was luck I guess...or chance, whatever you'd like to call it. But I wasn't putting myself in a position where I had information to make the choices. And I want them to be able to do that. (5: 41-51)

She later elaborated further on her hopes that her children will choose to use the knowledge that will be available to them to avoid having a baby with CF but she wants them to make this choice from a position of autonomy in which they feel they are supported regardless of the decision they make:

I'd first of all, like I said, like to get all the information in current research and what's happening. But I would really like them to know what it means to actually have a child who is...who has the disease. But I'd like them to know too that they have options along the way [...] they can do those tests early now in the womb [...] [s]o that they know if they're...if they've got the disease [...] then they have choices. And I would support them in their choice but I would feel very strongly that they shouldn't go ahead and have a child who they know has got cystic fibrosis. Very strongly. [...] But then I don't know that they're going to view it like that and they've got a right to view it differently but I certainly wouldn't be presenting an optimistic point of view. However, if they went ahead and had a child with cystic fibrosis then I would be very supportive and optimistic. But I'd want them to know that they didn't have to and that that's not a bad thing and I'd want them to have the support to get through making a decision like that and not feel bad about themselves or what they've done. (10: 34-53)

Here, Cindy touches on the cornerstone concept of Existentialism that, rather than absolute *Freedom*, knowledge and choice in fact bring a *Responsibility*-laden kind of

Freedom. This *Responsibility* chains the seemingly 'free' person and manifests itself as *Sartrean Anguish*. Cindy acknowledges that even knowing one has the option of having a genetic test brings the weight of *Responsibility* and the burden of knowing one has the *Freedom* to make a choice:

Choose your pain – how do you want your pain? Do you want it at this point or do you want it at that point? So as soon as you know, even in the knowing there's pain – pain about making decisions. (10: 55 – 11: 2)

And, because of this, she continues to emphasise how important it is that people have access to all the available information:

[T]hat's where the support and the information need to be around so that people can make those decisions one way or the other and live with the decisions that they make. (11: 11-13) [...] [I]t's about making a decision – not being positioned by lack of information or lack of support or biases. It's about making a decision and supporting the decision-making process for people. (11: 34-36) [...] [I]nformation is the crucial thing, I think, and the ability to make choices within the information that they've got. (12: 17-18)

She contrasts her own experience of being pregnant with her first child in 1984 with the experience that she considers will be open to her own children in the years ahead; her focus is on the power that she believes information confers:

I was positioned as still thinking, "Well, you either do or you don't. You run the risk. You don't...you can't find out if you're a carrier. All you can do is find out if you have a child who has CF". So there wasn't any room there to go anywhere beyond that. So unless you just didn't want to have children at all because there may be a risk, [pause] nothing you could do. (14: 15-19) [...] I'm just glad that now, for my kids, there's information. And that they've got power to make decisions and that it's not something that a family has to manage [alone]. (16: 5-6)

Yet, just when Cindy has persuaded us that 'things are so much better now', she astonishes us by declaring:

But I'm glad I missed out on having to be in that position – by luck, design, whatever – I'm glad I missed out on having to go through

that. Or having to make a decision. Or knowing that I was having a child, carrying a child with CF and having to make a decision about that. (16: 18-20) [...] That would have just been devastating. (16: 31) [...] And I'm glad my children [pause] won't even have to get in that position if they don't want to – because they'll know what their chances are. (16: 20-22)

Her 'ignorance is bliss' confession called into question the reliability of her conviction that information and choice are the keys to a person's ability to cope with the threat that the carrier risk poses entering into pregnancy. As a result of this, her each-way bet for her children cannot be paid because "know[ing] what their chances are" will not necessarily remove the need to test *in utero* if they wish to avoid the birth of a baby with CF, and it will not lessen the "pain" of the situation that this may throw them into.

The paradox underlying the insight that Cindy gives us into the value of information reflects the tension between the opposing schools of thought that it is 'better to know' or that 'ignorance is bliss'. The (un)wanted information occupies an *ambiguous* position within Cindy's life – on the one hand, she reifies what it will do for her children and, on the other hand, she vilifies the thought of the *Anguish* it would have created in her own life.

THE INFLUENCE OF OTHERS

There was an indication that other people, including health professionals, do openly judge parents and the decisions that they make around pregnancy and prenatal testing. At times, their judgements influenced the prospective parents' course of action; at others, it inspired rebellion; sometimes, it brought comfort...

Prior to finding out that only she was a CF carrier and not her partner, Vicki recounted a conversation that they had with a registrar at the hospital. In response to his advice that they would have a one-in-four chance of having a child with CF in each future pregnancy if it turned out that they were both carriers, Vicki explained that it was important to them to go ahead with testing because they would

probably decide against having more children in the face of such a high risk (10: 20-24). Understanding this as their viewpoint, the registrar made a remark which reflected a rather outlandish judgement call on his behalf that:

that sort of decision is quite harsh because now there's so much more that they can do for people with cystic fibrosis. (10: 28-29)

While it was certainly appropriate that he inform them of the changes that have occurred in the treatment and prognosis of cystic fibrosis in recent years – indeed, the context of the conversation demanded this – the occasion required that the information be given in a neutral manner that was not dismissive of their qualms about knowingly conceiving a child with a severe life-shortening disease. In this statement, he did not sufficiently validate their understandable fears about the distress and suffering that a disease like CF can bring to a family. However, Vicki said that in spite of this, had it turned out that they were both carriers she believes they would have remained steadfast in their determination not to risk CF in a second pregnancy (given that they both agreed they would not choose to test with the view to termination) (10: 52-56).

While Vicki and Tim maintain that the registrar's opinion would not ultimately have affected their decision, Stacy's own recollection is that she was influenced and guided by health professionals and the 'CF community'. Talking about her feelings after her first son was diagnosed with cystic fibrosis, Stacy said her initial thoughts were that she would not have any more children:

I thought "Nah, this is it, not gonna have any more" – wasn't gonna go through that again, in case I have another one. But, then I was talking to a lot of the nurses and a lot of the other mums and they said "Oh, don't be silly, you know, you can't not have a normal child, blah blah blah, you know. Take the chance". [...] [Y]ou can't let yourself not experience that as well. So then we decided that we would have another child – but I wanted to get tested...to see whether I was going to have a child with CF. (3: 31-43) [...] 'Cos I didn't want to have another child with CF. (3: 55)

It sounds like Stacy was highly influenced by the attitude that other people had towards her having a second child. One wonders if her resolve not to have a second child was indeed as firm as she represents it or if they were just her initial thoughts; a knee-jerk reaction that she was readily dissuaded from. One could speculate that she felt some level of moral doubt about whether she should have another child and that hearing other people's acceptance of that – even, their encouragement of it – made her feel okay about her desire to have another. Perhaps, for her, it was akin to receiving a stamp of approval of a kind.

What is so interesting here is the view that “you can't not have a normal child”/ “you can't let yourself not experience that as well” suggests that the other ‘CF mothers’ and nurses see having a “normal child” as a fundamentally different experience to having a child with cystic fibrosis; indeed, seemingly as a ‘must’, a fundamental need for any mother. Could it be that they feel a mother needs to have a child for whom she can project adult hopes and dreams onto; at least one child who will be around when she gets old? Or is it because of some antiquated notion of what this will do for Stacy's own sense of being **validated as a mother; as a woman; as a success?**

It is striking that Stacy recalls their attitude toward her expressed decision as one of “don't be silly” – this really belies the seriousness of the matter about which she was speaking and belittles the reasons and emotions underlying the rather understandable way that she was feeling. It is noticeable that she ends retelling what they said to her with “blah, blah, blah”, perhaps indicating that she felt at the brunt of an overwhelming, rather outspoken consensus around the idea that she should try to have a ‘normal child’. One wonders whether the ‘chance’ she was being encouraged to take was ever actually articulated or whether there is an unspoken understanding about the nature of this chance. Did the speaker mean she should ‘take the chance’ that she may have another child with CF or ‘take the chance’ that she may have to terminate if the foetus is affected? In making this statement there was little room for validation of her feelings as it seems there was

no consideration of her own set of beliefs in giving this advice in such a flippant, nonchalant manner.

JOSIE'S TALE: "OH, HERE I GO GETTING ANOTHER BLASTING!"

The interview with Josie reveals a double-edged encounter with the medical profession. On one hand, she had to battle a doctor who was aggressively directive in his counsel to her; on the other hand, she found professional genetic counselling staff to be a source of support and acceptance.

As stated in Chapter Three (see section: *Self-estrangement* (a form of alienation) versus *Authenticity*), a withdrawal from the 'they' has the potential to bring a person into conflict with others. Two years after their first daughter was diagnosed with CF, Josie and her partner decided to conceive again. Knowing they would not terminate an affected foetus, and concerned about the risk of miscarriage associated with chorionic villus sampling and amniocentesis, they could not see any reason to undergo prenatal testing. So began Josie's fight to follow through with this decision.

In an act of *Authenticity* and rebellion, Josie interpreted her situation in accordance with her own set of ethics and arrived at a perfectly logical conclusion based on that. Because of this, she experienced the wrath of the 'they' – **Thou shalt 'use' all scientific knowledge and technology available in the quest to eliminate imperfection of the human form** – via the less enlightened doctor she had the misfortune of having her initial consult with:

[T]he older doctor that we had at the [first hospital], he was horrified – absolutely horrified with us. He was so mad, with me, for going against any testing. He was so mad. He got me into the genetic counselling at the [other hospital] within like a day or two and he was so mad with me. [laughs at how mad he was with her] (18: 44-48) [...] I was like really distraught by him because I thought "Ooohh, why's he doing this to me?" I mean, being pregnant, your hormones are everywhere anyway! [...] [B]asically the doctor said to me: "Okay, you know you need to have these tests?" [mimics his low, grumbly,

clinical voice] And I said, "No, I don't want the tests". And yeah, he was just...I mean, he didn't yell or anything but he was quite cross with me, you know. He was quite angry with me. And yeah, he did probably think I was irresponsible, stupid, you know like, for not having them done. But I couldn't get it through to him that no matter what the tests were, I was going to have this baby anyway so I couldn't see the point of having the risk of miscarriage...having the tests done, if I was going to have the baby anyway. But he couldn't get his head around that. He wanted to get the tests done... (19: 19-36)

With great magnanimity, Josie said she could forgive the doctor for his attitude because of his age (she later gave some anecdotal evidence of her experience with other older doctors to back this up):

[H]e must have thought I was completely stupid or something but I didn't really blame him because he was an older doctor and I...the experience that I've had with older doctors is they remember CF what it was like like in the 80s or whatever when they didn't make five and ten [years old] and things like that so I sort of really wasn't too mad with him. I was mad with him but I wasn't mad with him because I thought, "Well he's stuck in the older generation of CFs, sort-of-thing". (18: 48-53)

Here it seems a possibility that Josie's superior knowledge allows her to show a generosity of spirit toward the doctor whom she perceives as 'out of touch' with the cystic fibrosis that affects today's generation of children. However, she admits that his behaviour did make her begin to doubt herself until her appointment with professional genetic counselling staff at another hospital; an experience she describes in overwhelmingly positive terms:

And then when I come up to Genetic Counselling [at the other hospital], they were just wonderful. Like I was fretting; thinking "Oh, here I go getting another blasting! Am I doing the right decision, you know, like not having these tests?" And I sort of thought to myself, "I don't really want to have this test. Why am I getting bullied into 'em?" And no, they were just brilliant. They just said: "Look, it is your decision. You know, which way, like if you do have the test, if you don't have the test – what's the point of having a test if you're going to keep the child anyway?" etc., etc., you know like...

They were so good up there. And yeah, I just walked out of there a totally different person than when I went in – ‘cos I was fretting, I was thinking, “Ohhhh no, another grilling!” But yeah, they were just what I needed. (18: 44 – 19: 7) [...] They just listened – I s’pose is probably the key word. And they could understand my point of view [...] You know, what is the point of having these tests? Yes, we’ll find out if she’s got CF or not. Yes, it will be a good thing probably to find out because when she’s first born, like for those two weeks, two or three weeks, before we get results back. But being pregnant, I was doing all the right things anyway so I don’t think I could have really done too much more. [...] [T]hey were just really good to me and just... It was so good: like I remember feeling...walking into there thinking, “Ohhh, here we go again – get another grilling”, and walking out of there so happy, like thinking, “Oh, finally there’s someone in this world that agrees with me”. Like, as in like, it was so different – I even told them that – it was so different like to what I expected. I thought they’d be on the doctor’s side for sure and they weren’t gonna be on my side and it was just lovely that... I think I even had a few tears up there ‘cos I was that distraught about coming there that day and getting bullied into these tests that I didn’t want to have done. And they were just so the opposite and it was really good so... And then, yeah I didn’t hear another word of it after that – didn’t hear nothing else about getting tested or anything. It must have been clearly in my file! (21: 11-36)

This is what a genetic counselling session should be – Josie came away feeling that she had been listened to, that her very real concerns had been validated, that her assessment of the risk as one outweighing the benefits was justified, and she felt free to make the decision that was right for her and her family within the confines of her own value system. She was empowered to rebel against those who would say that it was imprudent of her not to undergo prenatal testing, thus resisting an act of obedience and submission to the ‘they’.

Responsibility Constrains Our Freedom

WHEN TERMINATION IS NOT AN OPTION

One mother felt it would not be right *for her* to abort an affected foetus yet she sanctioned her own children taking this action in the future. For her, the context

in which this decision is to be made is important. In her circumstance, she did not feel it could be justified.

Josie refused to consider terminating her second pregnancy; she elected not to have a prenatal test for cystic fibrosis. While this 'choice' was seemingly hers to make – chorionic villus sampling would have been highly accurate in this case – her story does not reflect a decision based on free choice but rather on personal obligation and the duty that her and her partner seem to feel inheres in the role of being a parent. She positioned the feelings of her daughter, Diane (who has CF), as her primary responsibility:

I thought about, "Well, if we do find out these tests were positive and we've got another child coming with CF and if we abort him or her, how's Diane going to feel?" Like, say we don't tell her now...but family secrets always get out, like even when she's twenty [years old], how's she going to feel that we aborted a child because they had CF...because she had it? We couldn't do it to her. I would just think that she would feel like mud, like... I mean, she may not – she may think, "Thank you, Mum and Dad, for doing...making that decision – like not putting another child through what I had to go through". She may be like that but we don't know and we sort of always felt that taking it from her point of view and how she'd feel if she found out that we aborted a child with CF. And that stuck in our mind more: "If we ever wanted another child...we've done the CF thing – I'm sure we can do it again". And that's why we made the decision not to do anything [not to have a prenatal test]. (18: 33-44)

There were other factors that influenced their decision not to test for CF in their second pregnancy but Josie maintained that Diane's feelings were their main motivation for rejecting genetic testing technology and accepting that their next child may also be afflicted with the same illness:

There was a lot of factors. One of it was being scared of abortion – like I'd never had one and I'd heard all these horror stories. But I just couldn't see myself going and having an abortion. You know, I couldn't...we wanted this child. I couldn't... 'cos it's got something wrong with it, couldn't go to abort it – like it was against my grain, like it just couldn't happen [...] Diane was a strong part in it because I

just felt for her like – looking back, I was thinking like...if I had CF and I was twenty [years old] and I'd found out that my Mum got pregnant two years after I was born and aborted it because she had CF, it wouldn't make me feel very nice. It would make me feel like I was being a burden all these years and she didn't want another child with it. I mean, she may not think of it like that – she might think, "Oh, you did a good thing, Mum. Because you've got one, how about coping with two?" you know, she might...but it was just at the time. And...so yeah, we really did think of Diane in that way. And we really did think of...having the abortion wasn't the right thing to do. Like...not because I'm against abortions or anything like that. [...] I couldn't see how I could abort a child that we wanted – just 'cos it come a little bit earlier [than expected] – and it might have something wrong with it. (27: 12-35)

The insight given here into how Josie arrived at the decision she did confirms Sartre's assertion that "one makes a choice in relationship to others" (2004, p.362). While one could argue that she was totally free to choose other than she did (in fact, one doctor exerted enormous pressure on her to do so), in reality we see that she felt compelled to act within the strictures of being a mother. For her, this meant considering her daughter's interests as inexorably bound with her own – a continuation of "the intertwining and spreading of their ongoing coexisting" that began in a 'chiasmic relationship' when Josie still 'held' her as pre-infant in her womb (Wynn, 2002, p.9)¹⁵⁰.

As Josie considers the possibility that her children may one day face a similar choice, we see that she frames any potential decision of theirs in a different light to her own, attesting to Sartre's statement that "[c]hoice always remains a choice in a situation" (2004, p.362). Josie said the decision would be theirs alone to make:

¹⁵⁰ Francine Wynn delineates pregnancy as a "chiasmic relationship in which there is a simultaneous holding/being held of both mother-to-be and the pre-infant" (2002, p.5). She uses Merleau-Ponty's insights on embodied existence to do this, extending his phenomenology by arguing "that the origins of the phenomenological living body arise in the womb rather than after birth" (2002, p.14). Wynn also borrows his interpretation of the term 'chiasm' in *The Visible and Invisible* (1968, p.264-266) to describe a relationship that is characterised by "a mutual crossing over and withdrawing back into individual particularity" (Wynn, 2002, p.5). She hopes that understanding "this original relating" in this way will give us "a richer appreciation of the intertwining of later relationships" (2002, p.4), which is why I introduce it here.

*I would have to leave it up to them. Because it is a major responsibility. Yeah, it would be hard for me to get my head around it if they did decide to abort a child and not what I'd...like the opposite of what I did with [my youngest child] [...] But no, I would have to leave the decision up to them. Because they've lived through it as well, they might have had enough. They've always had to compensate – well, not always – but a lot of times had to compensate for Diane and things like that. And they may decide, "No, we're not going to have a child with CF. I'm not going to put my child through this" and things like that. I wouldn't mind either way like because they've already had to...**they've done their time**, you know what I mean? They really have so it would be up to them if they want to do it again or not. I don't know how Diane would feel but I'd say she'd understand. I couldn't see her understanding for me so much about aborting a child 'cos I know she...but I could understand her knowing that the brother or sister were going to have children. Because they'd understand that she knew that they've done their time as well. It's like if she got pregnant and she had a CF child coming, she might decide to abort it. Like: "Nah, I'm not having one! [said in a 'No way!' tone] I went through this – I'm not letting my child go through this". (25: 38-53)*

Upon prompting, Josie went on to clarify her introduction of the concept that Diane's siblings had 'done their time', saying "Meaning: living with it – living with CF" (26: 4). She explained that Diane's brother and sister have "just had to deal with so much", including being "shuffled" to different babysitters, learning to live with the times when "the household gets uprooted" because their sister is hospitalised for a two- or three-week period, and going to school after spending long evenings visiting her (26: 12-16). She talks of the complications that the need to take medication and be home to do physiotherapy introduce into a normal camping trip, and that "if they were all healthy and normal kids, we could do different things sometimes" (26: 27-37). She outlines all of the reasons why Diane's illness can mean all the children end up being late for school in the morning but adds that it is also about the emotions they go through and because of "a whole range of things" that they have 'done their time' (26: 37-43):

"Just, I feel like they've done their time; as in, they've lived it, they've...I mean, they haven't actually had the disease but they've

been so close to it and done sacrifices for it. Like...they've done their time, if you can understand what I mean? [...] They've made allowances for it. (26: 19-27)

Josie's words illustrate just how much a disease like cystic fibrosis becomes a disease of the whole family – everyone endures the upheaval of emotions that come with relentless treatments and hospitalisations. Because of this, she supposes that:

[t]hey might not want to do it [live with CF] again. They're still going to have – when they grow up – they're still going to have Diane. Fingers crossed. So they're still going to be – not so much living with it if they go live somewhere else – but they've still got her and they just might not want to do it all over again, you know like, for the rest of their lives. But, I don't know. I dunno. (26: 52-55)

The fact Josie sees the option of terminating a CF-affected pregnancy as acceptable in her children's circumstances but not her own reflects the concept of a paid debt in life, reminiscent of the Nietzschean notion that we stand in relation to our community¹⁵¹ as a debtor does to their creditor (Nietzsche, 2004, p.160) – her children will have 'done their time' (paid their debt) by the time they reach their childbearing years and, hence, having established themselves as *responsible* citizens, will have purchased enough 'credits' to allow them some moral leeway in putting themselves 'first' in their own decision-making.

We wonder if Josie feels she has not yet paid *her* own debt. Though perhaps this is irrelevant in her case given her focus on her daughter's feelings. Or is it perhaps that in her case the true debt that arises from guilt (Nietzsche drew attention to the fact that the German word for 'debt' (*Schuld*) arose from the word for 'guilt' (*Schulden*) and, hence, he implied the relatedness of the concepts) – guilt from 'passing on' a copy of the CF gene mutation to Diane and 'giving' her the disease – can only be relieved by demonstrating to her daughter that, no matter what, she would never have rejected her if she had had the choice and that she is not a burden to her parents.

¹⁵¹ And even to our ancestors and our gods/God (Nietzsche, 2004, p.178-179).

WHEN HAVING ANOTHER CHILD IS NOT AN OPTION

While it may initially appear that genetic testing technologies open up more options for couples at-risk of having a CF child, the narrative of one couple illustrates that once the values, perceptions and life experiences of people are taken into account, the options that are truly available to them become restricted.

Although it was not theorised in an existentialist way, as with Josie's account, the theme of responsibility also "emerged during discussions about reproductive decisions" in a qualitative study that examined the experiences of individuals who themselves have a genetic condition (Petersen, 2006). However, the theme of responsibility for 'others' was evident in the stories of those who had decided not to have children and who had "rationalised [this decision] in terms of their concern for future offspring". While this contrasts with Josie's story, it resonates with the sentiment expressed by Vicki and her husband, Tim, in this study.

At the point when their three-week-old son had been recalled for a sweat test following the neonate screen for cystic fibrosis, Vicki and Tim were told that he had been identified as having one CF gene mutation but that there still remained a chance he had another untested-for mutation of the gene, and would therefore have the disease. Even if he did not, this would be no guarantee that both Vicki and Tim were not carriers and, if they did both carry the gene, the implication would be that in each subsequent pregnancy they would face a one-in-four chance of having a baby with CF. Vicki spoke of their response to the registrar at the time he put this information to them:

[W]e said, "Well, you know, we would like to know because it would affect our choice". We probably wouldn't...at that stage, we had thought we couldn't have another child knowing that there was that high probability. We thought, "We've had Zane and, you know, that might just have to be it". [...] See, we wouldn't actually terminate once we were pregnant [and] I just don't think I could bear the thought of bringing a child into this world knowing that, you know, the quality of life that it had. I mean, I know that it's improved a lot and...but yeah, I guess we just thought, especially because we had

Zane... if we hadn't had any children at all, it would be a very difficult choice to make [...] but at least we would have had one child who was – I already know he was born healthy. (10: 20 - 11: 6)

Vicki later went on to explain why they could not bring themselves to terminate a pregnancy:

I don't think I could do it and my husband would be very much against it because one of his cousins, again, was told that their child would have a very high probability of having Down's syndrome and it was recommended that they terminated the pregnancy but the family went ahead with it and it was a normal, healthy child, so...yeah. (17: 11-15)

She revealed that she is relatively unimpressed by the current medical and therapeutic advances in treating cystic fibrosis and what it can do for the lives of sufferers, and it would seem this tempers any optimism she may feel about the improving prognosis for children and adults living with CF:

[A]s people keep pointing out, there are so many advancements in care of people with cystic fibrosis, but to this point, you know, apart from lung transplants, I mean, what can you do? It's not like there's a cure for the disease. [...] And how many people are lucky enough to get lung transplants and to actually have lung transplants that take? (18: 11-19)

We see that Vicki's concern for the quality of life of any future children she has and her part in being knowingly 'responsible' for that with the knowledge she now has, her lack of faith in what Technology will be able to do for CF sufferers, along with her and her partner's feelings and life experience around termination of pregnancy, leaves them with only one option ahead of them – not to have more children. So for Vicki and Tim, despite the existence of genetic testing technologies and despite the provision of information and counselling support, there is no *Freedom* in having to make a '**choice**' such as this because the power these things bring places *Responsibility* for the quality of life of their future offspring firmly in their hands – and this **compels** them to act as they do. They do not have the luxury of being able to say, "Let's just take a chance".

GENETIC INFORMATION AND ITS POWER 'RELATIONS'

The Choices of Others Confine Our Possibilities

Da-sein stands in subservience to the others. It itself is not; the others have taken its being away from it. The everyday possibilities of being of Da-sein are at the disposal of the whims of the others (Heidegger, 1996, p.118).

CLAIMED RIGHTS OF THE PARENT

It is not only the individual who has a vested interest in their own genes.

Sometimes parents claim a vested interest in the reproductive decisions of their own children.

So strong are Wendy's feelings that she would not be able to deal with one of her own children having a child with CF, she claims she will insist her children's partners are tested. Though she acknowledges that this sounds "selfish", she is emphatic that she could not deal with 'living with' the disease again:

[I]t's a horrible nasty disease [...] I can assure you I really don't want (as horrible as this sounds)...I don't want to have to go through it with my children with their children – I don't think that I could ever, ever have to go through that again, you know. To outlive my siblings is one thing but to outlive my grandchildren would just – for the same thing – would just, nooo, I think that that would probably just...that would just tip the time-bomb that stopped ticking away in the back of my head – that would just be...I couldn't deal with that at all. [pause...she becomes anxious] Ooooh God, perish that thought. That's why I want to find out because it's like I have to insist to my children that, "You have to have your partners tested": simple as that. Simple as that. I mean it might be for selfish reasons. They might think I'm selfish but I don't want to have to put up with that – I'll tell them straight out. I won't say to them: "Have your child with cystic fibrosis – don't bring it home to me!" I couldn't do that but, ooooh, I'd be devastated. I'd be absolutely devastated if I couldn't...I don't think that I could deal with that again. Chances are I'd outlive them... (22: 35-50)

Having suffered the worst at the hands of this “horrible nasty disease” already, it is clear that Wendy’s own experiences of the disease are impacting on her anxiety around her own children’s reproductive choices. Fear that she would not cope with ‘living with’ the disease a second time around and the fact that she would see herself as having to “go through it with [her] children” give us an insight into why she is **claiming the right** to attempt to influence the choices her children make.

PROTECTED RIGHTS OF THE CHILD

There were parents who showed a keen awareness of the fragility of the rights-in-trust of their children. They were very concerned not to influence what their children decide to ‘do’ with the carrier risk information and how they view it. One couple indicated they would delay imparting the information for as long as possible in order to protect them from premature worry or unhappiness.

Anne and Kevin said they would not seek to have their girls tested for the CF gene mutation – the thought had not even occurred to them until they received the letter about this research study, which then prompted a discussion between them on the issue (23: 13-18). They spoke about the outcome of that discussion:

Anne: [W]e’re quite happy and able to talk to them, and when they get to that stage of their life, um...

Kevin: ...they can make that decision.

Anne: Yes. I don’t believe we will try to influence it in any way, um... [...] Why would I want to impose unhappiness and risk and worry on them before they need to face that and they’re both people, I think, that will get to that stage and work it out for themselves. And I don’t want to come in and deny them any happiness that we’ve had but, equally so, should either of them want to talk about it there’s no problems there. No keeping it back [said quietly]. (23: 18-31)

Anne spoke of being glad herself that she had not known for years prior to her pregnancies that she was a CF carrier. Her view seems to be that the information should not be imposed on anyone and that it may be best to delay seeking it until

the point in time when plans for parenthood are coming to the foreground. She muses over this:

What if I'd had a blood test at twenty-one [before we got married] that had said – then – you're a CF carrier and you're this carrier and that carrier and that carrier, how much joy or happiness could that have robbed? How much worry could that have started at twenty-one? [...] And we weren't thinking about having children for a long period. (22: 20-27)

Her attentiveness to her children's right to an open future extends to their emotional wellbeing – their right to a future that is not laden with worry or concern before it needs to be. Her gratitude that “joy [and] happiness” were not “robbed” from her too soon means that she wishes this same possibility for her own children too.

At this stage, Anne and Kevin's children do not know about their one-in-two chance of carrying the CF gene mutation or even that their mother is a carrier. Consistent with Anne's philosophy, Kevin said they will probably not raise the carrier risk with their children until such time as they “announced that they were going to get pregnant” (25: 13-14). Anne speculated that “a natural conversation” arising out of the girls learning about biology and genetics at high school in the future may occur, and thinks this will trigger them to make the connection between their cousin's illness and their own carrier risk (25: 20-24). She continued:

I can't really imagine a conversation in which we would sit down and say “Kids, these are the risks of having children”. You know, it seems a very artificial and a negative...it's prejudging, you know, it's prejudging the information and I am very cautious about not laying anything on them or giving them any concern or worry about their lives. [...] I'm pretty confident that they'd make the connection and they'll come back to us – at which point I will be happy to provide the information. (25: 24-31)

In short, Anne is waiting for their approach which she says “doesn't feel like denial” (25: 36) or “any form of shame or deep concern” (25: 44), just the most natural way to relate the information in a way that will leave room for her daughters to decide

the level of significance it should have for them in their lives. This is quite different to the approach planned by some of the other parents. For instance, Stacy said that if the conversation arose with her daughter now, she would simply say:

"[Y]ou are a lucky one and you didn't have CF but you're just a carrier. So that means when you get older that you have to have a test done – to make sure that when you have a baby that it doesn't have CF." (18: 2-4)

She makes a number of assumptions about what action her daughter will take in response to the information she is giving her and, in this way, inadvertently imposes her own judgement about what the information should mean to her. The danger is that, without careful reflection on the way she conveys her message, she may foreclose some of the possibilities open to her daughter.

Anne's 'knowledge on a needs-to-know basis' approach illustrates a kind of pedagogic tact and thoughtfulness¹⁵², which draws from a recognition that her own choices may confine the 'possibilities' of her children. Thus, she demonstrates a desire not to constrain the *Freedom* of her children by presenting information too early or too formally:

Anne: *I worry if we sat down with our children now and posed [pause] that information, yes it would – they are very susceptible and they're making choices on their information on their life – it would be very easy to thwart their... Wouldn't it? [asking Kevin] Don't you think it would be easy to set up a condition of worry in them?*

Kevin: *Oh yes.*

Anne: *Because, sow a seed now that you don't really know how it would work out and [mumbles]... You have to be careful about doing that.*

¹⁵² Max van Manen defines pedagogy as "the activity of teaching, parenting, educating, or generally living with children, that requires constant practical acting in concrete situations and relations" (1990, p.2). He claims that pedagogic thoughtfulness and tact are essential elements of pedagogic competence and that this, in turn, relies on 'action sensitive knowledge'. It is his belief that phenomenological research, in particular, gives us this tactful thoughtfulness which he describes as "situational perceptiveness, discernment, and depthful understanding" (1990, p.156).

Interviewer: *Mm. So you think it's important the way you say it and when you say it?*

Anne: *[Replies in affirmation] The way you present it, the importance you give it, the implications, I mean...*

Kevin: *Oh, it's a societal process. Like it's about how society views these things. [...] So when you get to the point in society where it's not an issue for society and there's just information, then there's no harm in saying to the kids, "This is the information".*

Anne: *Yeah.*

Kevin: *And this is about: society doesn't yet value diversity. This is a diversity question.*

Anne: *Yes.*

Kevin: *And so, you know, once you get to the point where society values diversity, it's [just] a point of difference. (33: 43 – 34: 27)*

Anne is mindful of preventing the burden that worry can bring and this mindfulness is enacted through the way she plans to share the information with her daughters. She believes that how and when they tell their children of their carrier risk could potentially shape and influence the way they perceive the information. Anne shows a keen awareness of her children's right to an open future and does not want to risk compromising this. She sees herself as expanding their possibilities by leaving them the chance to explore the maximum range of reproductive options available to them. Kevin, however, focuses on the role that society has in shaping what being-a-carrier means to an individual. If there were no stigma attached to it whatsoever, he believes that telling their children would be unproblematic.

PARENTS KNOW BEST

Some parents feel they are equipped to make the best carrier testing decisions on behalf of their child – better, say, than those recommended by doctors or the generalist advice that medical ethics serves up; better even than the adult who

the child will one-day-become, it would seem. One mother used this to justify her demands that her children be tested while still only babies.

Rose's interview revealed that she vehemently believes the right is hers to have her children carrier-tested as babies. Her reasons for this were not necessarily convincing, although she herself seemed convinced by them:

I think we should really have a right to be able to test them – when, you know, to find out straight away. 'Cos what's not to say in a couple of years time, I might forget. Or something might happen to me and [my son] is put in care or something, and he had no idea. And you've got to start all over again. (23: 25-28)

Given her son's cousin has CF, it is extremely unlikely that his carrier risk could ever be 'forgotten'. One would have to say Rose is speaking from a position of *Bad Faith*¹⁵³ in using this 'reason' as justification for her demands that her children be carrier-tested as infants.

Rose revealed she intends to head back to the United Kingdom to live at some point but, believing it is too hard to get testing done there, she indicated she will initiate having her newborn baby carrier-tested at his 'six week visit'. On the possibility that the doctors in Australia also may not agree with her desire to have him tested at such a young age, she replied:

[I]f they said "No, I couldn't get him tested because, you know, let it be his choice when he's older", I'd freak – I'd fight that. As I said before, I'd rather know so I can educate him growing up [...] to the fact that he has this...faulty gene... (22: 31-39)

It could be inferred from her use of the word 'educate' that she sees herself playing a major role in how her sons understand and adapt to the genetic information that she has taken it upon herself to investigate and present to them. Her stance is diametrically opposed to Anne's, who is extremely concerned not to influence how her children perceive and respond to similar information.

¹⁵³ See Glossary of Terms to review the meaning of this existential concept.

On the subject of having the little boys carrier-tested as babies, Rose's own mother had said earlier in the interview that this would prevent the situation Rose had found herself in from occurring over again:

[S]he didn't know at the time she fell pregnant with [her son] whether she was a carrier or not. Whereas, with [her son], he can be brought up all his life now to know that that danger is there. So it's something that he's got to think about when he starts going out with girls and that sort of thing but "hey", you know, "there could be a problem here, I'd better...", you know. Whereas if he doesn't know, that's how a lot of things get passed on because it's not found out early enough and they don't know, don't take the precautions. (9: 27-33)

Rose agreed that her son is in a much better position than she had been (16: 54-56), explaining why:

[W]e knew nothing about it before. I didn't grow up knowing about it. It just hit me at an age of 23 [when her niece who was diagnosed with CF was born]. But whereas now, I can tell [my son] all the way through – there's not going to be a shock or a...he's not going to worry about it. (16: 49-52)

Being able to bring her son up knowing that he carries the CF gene mutation is important to Rose and her mother, who advocates routinely testing for the carrier state, as well as the disease, during the neonate screen:

I can't see why, when it is so widely spread these days, [...] why they cannot, as well as testing for it, while they're testing for it, why can't they not just test and see if you're a carrier, and inform you? Right from the start. I mean you know how to handle it then the rest of the child's life. (16: 16-23)

In saying this, Rose's mother does not seem to look ahead to see the child as an adult who will one day have the capacity to make his own decisions and "handle it" himself. Neither Rose nor her mother show any realisation that their actions/attitudes will limit the reproductive possibilities of their (grand)children in that the option of not being tested – not seeking the information – will be closed to

them. In fact, they would be hostile to the notion that it was the children's right to have this option remain open to them.

Throughout the interview, Rose also did not waiver from her belief that it is best for her children to be tested as infants as opposed to leaving it until they are teenagers in order that they be left to make the decision for themselves. While speaking specifically about her older son, she gave a number of reasons for this:

Rose: I've got so many years now to work up how to go about it, how to tell him and explain to him the consequences and everything. See – at fifteen and sixteen [years of age] – by that time, 1) it could be too late... (I hope it's not!) [...] But, you know, by then it may be too late [sounds anxious about leaving it that long]...and it would be harder for him to understand – [a quick thought] you don't want to listen to things like that...he might get embarrassed about it...

Mum: He'll say, "There's nothing wrong with me, I'm fine, I don't feel sick", you know...

Rose: And, you know, a fifteen-year-old boy...to try to take him to the doctor to have a blood test done for this gene that when he has... [...] [H]e could feel embarrassed about it; he could feel awkward, uneasy; he might not understand...what it's all about. Whereas if he knows... I know now – I can explain to him, as the years go by, more and more about it. (9: 44 – 10: 8)

In fact, there are years between now and when Rose's fears that testing will be left "too late" (9: 46) need to be invoked. It is clear that Rose feared her son would be resistant to carrier-testing as a teen and she attests to averting this scenario by seeking testing at a time when he was unable to be a part of the decision. Because she believed it was necessary to make this choice on his behalf as his parent who knows what is best for him. She consciously seeks to limit the possibilities available to him as a potentially 'resistant teen' in case he does not choose as she would like him to.

Information Custodians in the Family

WHAT PARENTS ARE SAYING NOW

In families where an instance of the disease is present, the 'moment of disclosure' to a child of their carrier status is unlikely to be a 'moment' at all. In many cases, it will be a gradual disclosure of increasingly sophisticated and detailed information that is more tailored to the individual over time. However, this may not be the case in families that have been untouched by the disease, where it is less likely there will be a trigger for 'the conversation'.

Tanya's daughter was picked up as a CF carrier in the neonate screen. She thinks "a recessive gene thing" is quite "complicated" to explain to a child and for this reason she would not talk to her four-year-old daughter about it while she is still too young to understand – it would not be "useful or serve any purpose [...] except to confuse her!" (14: 27-36):

We haven't said anything to her. She wouldn't understand it anyhow. But I mean, I guess when she gets older, we will tell her about it (12: 45-47).

Another mother whose baby was identified as a carrier in the screen predicts that it would be "frightening" for her son to hear that he is a carrier prior to learning anything about genetics, especially if he does not understand "carrier status as opposed to actually developing a disease"; she thinks "it would be a concept beyond him" (Vicki, 18: 29-31). It sounds like these mothers will not raise the topic of their children's carrier status at all until they believe they are old enough to understand the concept in its entirety. With no instance of the disease in the family, there is no reason why 'the conversation' will be forced before they judge their children are ready to fully comprehend the information.

According to Rose, it is the parents who are in the best position to judge when this is:

[Genetic information is] best coming from the parent, just...they know when the child's ready and what the child understands and what the child doesn't understand. Not [...] the doctor. (23: 50-52)

However, Rose herself proved that parents can get it wrong if they 'jump the gun' by saying too much too soon:

I have sat [my six-year-old son] down and said about the carrier thing – this is when I thought that it's too early – was, he said something like, "Will I die?" [sounds like she was shocked by this] And I'm like "No! No, of course, you won't!" And I think that's when I thought, "Well, it's scaring him a bit – too young". (13: 10-13)

This conversation was precipitated when she was trying to explain his cousin's illness to him. Rose presumed he misunderstood what she said in relation to his carrier status, concluding that he has "got this thing as well" (13: 21).

Her son already asks a lot of questions about his cousin's illness – what is it, can he get it (10: 40-42)? Notwithstanding the prior mishap, Rose tries to explain "in his terms" as best she can – i.e.,

"When you get older, [...] you're gonna have to be more careful when you have your little boys or little girls" (10: 24-25).

She says talking about genetics is "a bit complicated for him at the moment" (11: 9-10) and indicates she will give him more details about his CF risk "shortly – within the next few years" (11: 14). Her mother adds:

It's basically just a gradual thing, you know – each year you say a little bit more, and a little bit more, and a little bit more, and then at some stage before they get too old, they can see for themselves the whole picture – they can put all those bits together themselves so they understand. (11: 18-21)

We see that in a family where the illness is present, it is anticipated that a series of conversations will occur before the child's genetic information is fully **released to** them ... **from the custody of** their parents.

Josie's six-year-old daughter has an older sister with the disease. While Josie states she has told her she is a carrier – explaining, “You’ve got my gene which is the same number as me” (17: 42-43) – she says:

[S]he wouldn't have a clue what it means – she's only six. We just said, "When you meet up with somebody and have babies, you might have one with CF", and we just said, "But don't worry about that now". (17: 29-32)

In response to this information, she said her little girl “didn’t blink an eyelid”, which she attributed to the fact she is “too young”; “she doesn’t know” (17: 46-47). She explained this conversation had probably arisen “‘cos we do as a family talk about quite a few things and it flows on” from there, and that it was probably prompted by her daughter asking, “‘Why haven’t I got CF?’ or something like that” (17: 39-41). She continued:

I didn't want to overwhelm her and get her into distraught mode [...] [S]he helps me with physio and things like that so (well, her way!) [we laugh]...and you know I could have said to her, "You might need to do this [physio] one day". But I don't really want to fill [her and her older brother's] head with that, you know what I mean? Like it's so hypothetical at the moment. Like, you know, we're talking years away. I don't want that to be sitting in their minds. (17: 37-54)

Like Anne (see section: Protected Rights of the Child), Josie is mindful about not causing her children to worry prematurely about the risk that they too may have a child with CF. Her focus on the ‘work’ aspect of being a ‘CF parent’, rather than the emotional side of it, may be her way of managing this. We both agree that her youngest daughter probably perceives physiotherapy, etc. as a normal part of parenting anyway because she has witnessed her mother ‘at work’ at it her whole life.

Cystic fibrosis was always openly discussed in Cindy’s household too. This gave her children, at risk of being carriers themselves, a chance to come to understand the impact of the disease on their family:

[W]e talked a lot about my younger brother and there were lots of photos and my mother would talk about him and I would talk about him. [...] So they were aware of who [their uncle] was and why he wasn't here and...so we talked about cystic fibrosis and what it meant and what his life was like and how he died at nine and had to have all this physio [...] So they knew what it meant in the sense of from our point view. (6: 49 – 7: 1)

In addition to this, Cindy shared a piece she wrote about her late brother with her children so it gave them “a bit of an insight – not into the disease, but the impact of the disease” (13: 30-31). Her children also went along to the hospital when their mother and other adult relatives were tested “so they were kind of part of that process as well – taking the blood and all of that” (7: 14). Because of the family’s inclusive approach to sharing **information generally** about the disease, there was never a need for one conversation – one moment of disclosure – in relation to **genetic information**. Their coming-to-know of their at-risk status for being carriers was a process that occurred over time – a kind of knowing without telling. The familial aspect of their relationship to CF has been emphasised, rather than the genetic (or not) aspect. It is possible this will be reversed later if and when the children seek a genetic counselling consultation. According to Armstrong *et al.* (1998), the process of geneticisation that is enacted through the structuring of a typical consultation serves to “[entangle] the individual in the genetic identity of others” (p.1658), thereby “[locating] the patient in a genetic space and [affirming] the importance of genetic relationships” (p.1655).

SILENCE WITHIN THE FAMILY

There were a number of families where a silence existed around the knowledge that the disease or the gene, as the case may be, was present in the family. The reasons for this were not always clear but superstitions and taboos figured amongst them. Frustration was expressed by two mothers – one who felt her son’s CF was partly attributable to the silence and another who felt genetic information relevant to her and her partner would not have been forthcoming if they had not raised the matter themselves.

Henry professed that he finds it difficult to speak with his adult son about their shared carrier state because

I don't have much experience in that way, you know, to talk about sickness and all that. (11: 40-41)

He added another reason for avoiding talk about it too:

I think we sort of people, we are like maybe happier things to remember on, you know. Like these things is hoping not talking about it, nothing is coming out of it as well, you know. (sic)¹⁵⁴ (12: 32-34)

In his idiosyncratic way, Henry is saying that they tend to focus on the positives – the “happier things” in life. The explanation he gives for not discussing the issue borders on superstition, implying that he does not wish to tempt Fate by giving too much attention to the fact the CF gene mutation is in the family – that by not talking about it, nothing bad will come from it!

Stacy, whose son was born with cystic fibrosis, said that her husband’s family did not talk openly about the illness that claimed his cousin’s life many years ago. Despite the fact that they were of a similar age and “really good friends”, her husband was not aware that his cousin had CF until they were in their mid-teens (25: 32-37). She figures it was “a taboo-ey thing with them” (25: 31) that no one in the family used to discuss or ask any questions about (25: 37-38). Stacy feels “a little bit cross” now that this was the family’s attitude –

not being more aware of knowing that their cousin had cystic fibrosis, and not being more aware of, you know, them being tested and bringing it out into the open. (22: 45-48)

She feels that “maybe this could all’ve been avoided” (25: 41) if her husband had been aware of his CF carrier risk and “then [she] could’ve got tested” (22: 48-49) – that it was “a little bit careless on their behalf as well – that they didn’t do anything

¹⁵⁴ English is Henry’s second language and he is not entirely proficient.

about that" (22: 51-52). She says the family may not be the only ones implicated in the 'silence', however – that "it could've been the doctors' fault by not saying to them you know you should all get tested" (23: 10-11).

What Stacy forgets here is that the carrier test for CF did not become available until the year after her son's birth – no carrier testing was available when her husband's cousin was alive so it could well be that any 'education' the family received about the genetics of the illness was limited to the fact that a moderate risk existed that the disease could strike the family again. Other than raising the family's awareness of this, nothing more could be done at that point. The other thing Stacy has overlooked is the fact that the CF mutation in her family is not one of the common mutations that is tested for when a person with no family history of the disease undergoes a 'carrier test'. This means that had her husband been aware he was a carrier and she presented for testing, the CF gene mutation that she carries would not have been identified and she would have been advised that her risk of being a carrier was highly reduced. The birth of her son with the disease would not have been averted. Still, in her mind, the perception is that all her possibilities were not left open to her which leaves her feeling that her *Freedom* was diminished. One can see how a lack of understanding about the genetics and history of cystic fibrosis leave the family's silence open to blame in this instance. And because this blame is something she is reluctant to voice, and because the family is not talking openly, it is unlikely they will ever know that this second tragedy in the family was not preventable within the confines of today's testing technologies.

Ironically, even with Stacy feeling "a little bit annoyed that they were so blasé about [having CF in the family]" in the past (23: 1-2), she has bowed to the wishes of her mother-in-law and allowed the silence around the disease to continue into this next generation. Of her son's illness, she says that apart from one cousin who visits him in hospital,

all the other cousins don't know anything – don't know that he's got cystic fibrosis. I got told [by my husband's mother] that I wasn't allowed to tell 'em... 'cos they might treat Bradley differently. [laughs

like 'how stupid is that'] [...] "They don't need to know all the nitty-gritties" [laughs]. So they don't know exactly what's entitled (sic) with cystic fibrosis – they just know that he needs to go into hospital and stuff like that. (26: 43-51)

This is concerning because some of these cousins are adults already and may not be aware of their carrier risk. She expresses her thoughts about this:

I think it's quite childish really. But they seem to think that if we tell them all the nitty-gritties that they might treat Bradley a little bit differently. I don't know – they might not throw them over his shoulder (sic) or play footy with him in the backyard. (27: 5-7)

We see that she allows the family silence to be perpetuated despite resenting it herself. She allows others in the family environment to dictate how she lives with her own son's illness, even privately. Can she be getting the support she needs here?

Two of the couples broke a silence they were unaware existed in their respective families by raising the issue of carrier-testing with relatives. Rose and her fiancé were "glad" they spoke openly with his parents about his decision to be carrier-tested because otherwise they "may never have known" that "his cousin has it"¹⁵⁵ as well" (18: 23-24/ 17: 39-40). Importantly, this meant that he could be tested for the mutations specific to his family as well as the more common ones (18: 25-32). Vicki and Tim had a similar experience with his family. It was not until they told them the news about their newborn son's carrier status that his parents mentioned that one of their nieces also has one copy of the gene (3: 6-10). Vicki said she found it "quite frustrating" that the gene was already known to be in the family but no one had told them about it (although she did not express this to anyone) (4: 7):

I believe that people should be aware of that sort of thing – that's why I have no concerns about talking about cystic fibrosis because people, I think, don't understand what cystic fibrosis is. [...] [I]t only came out because we thought...you know, Tim and I discussed it

¹⁵⁵ It was unclear here whether she meant that the cousin has CF or is only a carrier but I think it is most likely she has the disease on the basis that Rose implied there was more than one CF mutation present in her fiancé's 'family'.

between ourselves. [...] [W]e thought that we should tell his side of the family, yeah, and then when we found that out, I was... [pause] It would be nice to know. (4: 7-14)

So where there had previously been silence, parents were fairly forthcoming with genetic information once it was known that the gene was also present in their children's partner's family.

Silence existed for a different reason in another family; this time it was in relation to a neonate screen result. Leanne and Rob did not tell anyone in their family when their baby, Amy, was recalled to the hospital for a sweat test. Leanne explains why she needed privacy at that time:

I didn't want to worry people if it was all going to be okay. So, I mean, I know I already got myself into a worried state but if other people around me were already...were getting worried as well and were saying, you know, "Everything will be okay" or, you know, and ringing up and finding out, that just would, for me, I guess again it was sort of a protective thing for myself, I guess. I would've...yeah, I think it just would have made it harder if I knew everyone else knew. (7: 30-38)

The extended family still do not know that Amy and her father are carriers but Leanne and Rob understand this has implications for them (7: 43-47). They plan to tell Rob's sister so she is aware of her own carrier risk but have hesitated to do so, partly because there may be "a bit of a backlash" (11: 32-33). Leanne thinks the family might ask why they did not tell them about it at the time so they could offer their support but she says they have always been a couple who "tend to deal with things ourselves" (11: 33-39). She is not at all worried that "everyone will know" their daughter is a carrier,

[i]t's more that everyone will know that we didn't tell them. [laughs] That's more of what is probably like the issue (11: 47-49).

What we see here is that the possibility of protecting one's own right to privacy, and not subjecting this to the scrutiny of others, is severely restricted where a

person feels a responsibility to share their genetic information with other stakeholders in the family:

I guess we could've not been tested and then just not passed it on but that doesn't seem like the right thing to do and I think if you, you know, if you've got the knowledge, they should, you know, they have the right to know. (9: 53-55)

As de Beauvoir says of the existentialist ethic:

What makes the problem more complex is that the freedom of one man almost always concerns that of other individuals. (de Beauvoir, 2004, p.424)

The *Freedom* of Leanne in this instance would mean that Rob's sister remains unaware of all possibilities open to her to minimise her chance of having a child with a genetic illness and this would "not seem like the right thing to do".

LEANNE'S CALL: "THERE'S NO POINT TELLING THEM IF THEY'RE NOT HAVING KIDS"

Leanne and her husband are the custodians of genetic information which affects his whole family but they only see a need to share it with stakeholders who are planning to have children. Their initial silence about it means it has now become a slightly delicate matter for them to bring up.

Leanne recalls that Rob "didn't really see the need" for them to undergo carrier testing when their daughter was identified as a carrier but he had a change of heart when he considered that his sister may need the information if she ended up deciding to have children (14: 46-52). Despite finding out he does carry the CF gene mutation, and there being "nothing really stopping [them]" telling his sister about it (10: 27), he has not yet advised her of this. He said he was "ready to tell her straight away but for one reason or another we didn't get around to it" (10: 25-26). Even though he has put off having this conversation, he does not anticipate it being hard to tell her and he thinks "she will be able to cope with that alright" (10: 31/39).

Leanne reminds Rob that they did not mention anything to his sister initially because they saw “no point” if she and her partner were not discussing baby plans at that time:

*I think the main reason you haven't told her was that issue that we didn't ever know that she wanted children. It wasn't until recently and her spending more time with Amy that that whole issue had come up. [...] [W]e thought maybe that she would but it had never come up so I think – well yeah – that's probably one of the main reasons why we haven't sort of...we hadn't said anything, because we thought “**Well, there's no point telling people if they're not having kids**” but we probably could have easily just told them then.*
(10: 46-57)

Rob's words are confirmation that he does have definite plans to tell her:

[M]y brother is not likely to have kids, for example, but my sister is – so there will be a time where I'll definitely tell her so that she perhaps can at least have the test beforehand, as perhaps we might have had that been available. (8: 5-8)

In saying this, however, he reveals a level of discrimination as to who needs to know the information and it is strictly based around who may need to use it. A judgement is made that only those with a **use** for genetic information have a fundamental **right** to it.

CONCLUDING REMARKS

Through “[t]he processes of exploring the family tree and mapping the genetic links” of the client, it has been found that the genetic counselling consultation actually serves to reinforce the notion that “identity [is] located in genetic make-up” (Armstrong et al., 1998, p.1657). In their study of these processes, Armstrong *et al.* (1998, p.1657) remarked that “[t]here was never discussion of ‘who you are’ as this was pre-given by the density of the genetic map”. They noted a process of geneticisation occurring, whereby

patients are at once incriminated within lines of inheritance and have the responsibility of that problem dissipated in the web of consanguineous relations that they share (1998, p.1658).

Overall they found that,

[d]espite its name, the genetic counselling consultation is an important mechanism in the process of revealing and consolidating an identity in a web of genetic connectedness (1998, p.1658).

In other words, the consult itself (more so than the genetic information) serves to provide the client with a genetic identity. In light of the findings in this chapter, this is not necessarily desirable. We have seen that people evaluate genetic information differently. Some do seem to integrate knowledge of their carrier status into their sense of who they are and, when they do, they speak of feeling stigmatised; of having 'something wrong'. It seems the client who views their positive carrier test result as a piece of information, rather than identity-defining, may have the healthier outcome. If the carrier 'state' is felt to be one of **knowing**, not **being**, it does not become a part of one's identity. When Armstrong *et al.* say the genetic consultation "[reveals] an actual identity in the place of a previously presumed one" (1998, p.1653), they echo the sentiments of the positivist program – that the truth idly sits in the world, in our bodies and, now, in our DNA, awaiting discovery. I contend that the genetic counselling consultation works to **produce** a genetic identity that 'does-not-have-to-be', rather than to **reveal** an identity that 'is'.

The focus on information, as seen in the data, reflects the technicity of our time and our place. As we have seen, those who know genetic information do not necessarily experience unfettered *Freedom* to act in relation to it as they wish. They exist in relation to others: they may be influenced by them; they may face pressure from them; they may feel supported by them. The Other has 'an indubitable presence'¹⁵⁶ in the genetic counselling/testing context.

¹⁵⁶ Here, I am inspired by Jean-Paul Sartre (1958, p.275).

With the knowing comes *Responsibility*. This too constrains *Freedom* in terms of making decisions around pregnancy and prenatal testing. In particular, those who know genetic information feel *Responsibility* towards existing children and also towards future children who, as a consequence, may never be born; never be conceived. Sometimes, due to their own values, perceptions and life experiences, their 'free choice' is narrowed substantially. With the knowing may come less *Freedom* to act as they will; less chance of 'taking a chance', 'come what may'.

Those who know genetic information become its custodian. They have decisions to make. They differ as to who they think has a right to the information, how much they talk about it, how they talk about it and when they will talk about it (this latter point will be discussed in the next chapter in the section related to Timing). They have power to impact on the possibilities of other stakeholders, particularly when these stakeholders are children. The *responsible* parental custodian realises this. As the likely deliverer of the information they have the potential to influence the way their child evaluates and responds to that information (because many do have hopes or, at least, assumptions about how their child will act in relation to it). We see that some parents are not mindful of this; some claim the right to attempt to influence their child; some are highly concerned not to prejudge what the information should mean to their child. We sense that the latter kind of parents will work hard to avoid compromising their children's right to an open future. This is '**caring for**' the genetic information responsibly.

Those more keenly aware of the fragility of the 'rights-in-trust' of their children are likely to find being sure of how and when to release the information more difficult; more fraught with danger; more dilemma-ridden. This is because they realise that they can violate these rights now in ways that will cut off the possibilities of their child. Those parents who show a consciousness of not wanting to foreclose any of their children's reproductive possibilities, evidenced by their troubling of how to tell and when to tell, see this as a delicate and sensitive matter; one to be approached with care. **This is pedagogic thoughtfulness-and-tact-in-action!** And it is properly governed by a sensitivity to their children's right to an open future.

CHAPTER SIX: TRANSCENDING GENETIC SITUATION

INTRODUCTION

The *Ambiguity* of the given (our facticity) and the taken (our transcendence) of our situation pervades our individual and social lives¹⁵⁷ (Flynn, 2006, p.66). With the advent of Technology, our genetic situation has now become *ambiguous* too. The genes we had no control over being given to us are a 'given' of our genetic situation, whereas the attitude we have towards this, as well as the minimal control we now have over the genes we pass on, are the 'takens' of our genetic situation. Over our **facticity**, we are **powerless**. But through **transcendence**, we can feel **power** over our situation. Hence, how we face up to this *ambiguous* aspect of our existence would seem to be key to how well we cope with it.

Sartre, echoed by Camus and de Beauvoir, said that because we can always transcend our facticity, we are 'without excuse'. Best outlined in Camus' *The Myth of Sisyphus*, the pessimism commonly associated with Existentialism actually "harbours a deep, if limited, hope" (Flynn, 2006, p.49). This Hope, within an otherwise *absurd* existence¹⁵⁸, is evident in some of the stories shared by the participants in this study. How are people living-with genetic information? How do they cope with the fact that they had no control over what genes they received and what genes they have passed on to their children? How do they look to control a situation over which they have had very little control?

This second analytical chapter considers the ways that people attempt to transcend their genetic situation.

Through recognition and willed acceptance of their facticity, people manage to live-with their genetic situation. We see this act of *Revolt*¹⁵⁹ enacted in various ways.

¹⁵⁷ A discussion of this was presented in Chapter Three (see section: *Ambiguity*).

¹⁵⁸ A discussion of this was presented in Chapter Three (see section: *Absurdity and Revolt*).

¹⁵⁹ This existential concept was discussed in Chapter Three (see section: *Absurdity and Revolt*).

People may find refuge from an uncertain future by living in the here-and-now. Concerns about genetic threat may be held in abeyance to be faced another day; decisions about when to reveal genetic information left for another time. When a close encounter with genetic threat has passed over an individual, they choose to feel lucky about the now-intensely-felt contingency of their existence, rather than *nauseous*¹⁶⁰. Many look to transcend the facticity of their genetic situation through what could be termed ‘**technological transcendence**’ – i.e., learning what they can about genetics, ascertaining the options available to them and accessing genetic testing technologies. While parents recognise and accept that they were powerless-to-control which genes they passed on to their children (this being their facticity), they focus instead on the power-to-control that they believe knowledge of their children’s carrier status will give them over their future (this being their transcendence). This, to them, is being *responsible* with their genes. Through deliberate choice, they believe they can rise above the *absurdity* of their genetic situation.

LIVED TIME AND TIMING

Time is of the essence. We are fundamentally time-bound beings. Unlike measurable, ‘clock’ time, lived time is qualitative: the ‘not yet’, the ‘already’, and the ‘present’ differ among themselves in meaning and value. (Flynn, 2006, p.8)

Learning to Live in the Present

STACY’S ADVICE: “WHY SHOULD YOU WORRY ABOUT SOMETHING THAT’S GOING TO HAPPEN TWENTY YEARS DOWN THE TRACK?”

The existential notion of lived time¹⁶¹ provides an elegant way in which to discuss how Stacy faces the dual task of parenting a child with CF and a child who is a CF

¹⁶⁰ This existential concept was discussed in Chapter Three (see section: *Nausea*).

¹⁶¹ The concept of ‘lived time’ refers to the lived experience of existential time and was mentioned briefly in a footnote in Chapter Two (see section: Reconciling the Christian and the Atheist Existentialists). It alludes to the “qualitative and personal dimension [of] the phenomenon of time-

carrier. Lived time can be used as a conceptual tool to offer one interpretation of why Stacy insists it is best to focus on today and not worry too much about what the future is going to bring. We will see that she has *learned* to live in the present.

After her first child was born with cystic fibrosis, Stacy chose to have prenatal testing at twelve weeks into her second pregnancy. Of interest to health care professionals in terms of how they convey 'news' to their clients is Stacy's account of how the information that her daughter is a CF carrier was relayed to her:

[T]hey actually rang me up on the telephone and they said to me, "I've got some good news and I've got some bad news for you" and I said, "Oh, what's that?" and they said that "your child is a carrier of cystic fibrosis but she hasn't got cystic fibrosis" and I think that I was just so overwhelmed with the fact that she didn't have cystic fibrosis, having...being a carrier didn't really... (4: 14-18) [...] I didn't even think about the 'bad news', I was just so over the moon that it was um [pause]... good news – that was good news to me. Just the bad news part of it didn't even enter my head. (5: 42-44)

Her words reveal that finding out her foetus carried one mutation in the CF gene paled in significance to finding out that she *did not have* two mutations:

I was just relieved. I just...it didn't even, I don't know, it just didn't even enter my mind about her being a carrier – I just was so happy that she didn't have cystic fibrosis (4: 40-41) [...] [I]t was a relief (4: 49). [...] I was just happy. I didn't even think about anything else. (5: 4)

Even nine years on, the knowledge that her daughter, Kristy, has one CF mutation has never even been a blip on the radar of everyday concerns for Stacy. Stacy does not believe that Kristy's carrier state has had any impact on her life so far and she is adamant that it has had no influence over the way they have parented her (15: 6-38). In fact, when asked how she felt about her daughter being a carrier, Stacy

consciousness", not to the dimension of time that we measure quantitatively in minutes and seconds (Flynn, 2006, p.5).

'confessed' that "to tell you the truth, I've really never thought about it" (1: 11-12).

She later added:

To be honest, it doesn't really – at the moment – mean anything at all...to me. I guess [thinks]...that's something I'm going to have to think about when she gets a little bit older. (6: 7-9) [...] [I]t hasn't affected me at all that Kristy's a carrier (15: 27-28) [...] [H]er being a carrier hasn't even entered my mind. (15: 38)

In some ways it seems quite surprising that she has never worried at all about the chance, albeit a small one, that her daughter may have a CF-affected pregnancy. At the same time, it is overwhelmingly clear that her son's health is her pressing concern for now, while any possible future pregnancies on her daughter's part are still years away. Stacy does not think too far ahead into the future. She focuses on the here-and-now.

Listening to her speak of her experience of living with a child with a life-shortening chronic illness, it becomes evident that living in the present is a way of being that she has found allows her to cope with the uncertainties which enshroud the future of her family. She reflects that the expectations she had for her son's quality of life when he was first identified as having cystic fibrosis turned out to be overly pessimistic and that "it's not as bad as what the doctors say it is" (2: 30):

Ohhh I was devastated. I thought that he was going to look different, that he wasn't going to be able to play football or soccer or eat normal and stuff like that. That's about it. [nervous quiet laugh] I just thought that I would have a really sick child on my hand. And he was gonna die really young. (2: 14-17)

Her son's diagnosis propelled her thoughts to the future, away from the seemingly 'perfect' baby in her arms. Her initial outlook on the years ahead was one of a bleak future for her child. She feared he would be different from others and her focus seemed to be on overt differences such as his appearance, his sickliness and his inability to join with other children in playing sport. The lesson here was that living in the 'not-yet' meant that she wasted a lot of energy worrying about her son's childhood, which in the end she feels turned out to be relatively normal. In the face

of her catastrophising, things turned out much better than she imagined and so she learnt to shun thoughts of the future and become absorbed in the present – over the course of his childhood she became immersed in the everydayness of living with cystic fibrosis.

When asked about her perceptions of the disease now, in terms of its severity, the burden it places on a family and how much it affects a child's quality of life, she finds it difficult to answer because she perceives that it impacts on some families harder than others:

[I]t's hard to say because Bradley's always been really well¹⁶² so it hasn't really been a big burden – but I know other families, when their kids have been really sick continuously and in hospital every three or four months, so I guess that I'm just, Bradley and us are just one of the lucky ones. (2: 36-39)

Stacy then reveals that despite their 'luckiness' so far she is caught between feeling reassured by their luck, and hoping that this will remain with them throughout the disease process, and a knowingness that the odds are against this happening (without going so far as to intimate that with CF it is an impossibility for their luckiness to continue):

I don't know if [our luck is] going to change – which it probably will – and that's always in the back of my head too. But you know he will get sick. (2: 49-50)

Her sense of security is never something in which she places all her faith for she admits that thoughts their luck cannot last are constantly percolating in the back of her mind. The fact she relegates this knowing to the "back of [her] head" indicates she is happy to allow herself to feel secure in the present for now. As she expresses her feelings over whether their luck will continue, I note that her twice-revised sense of security reveals the conflict going on in her mind about what their luck now implies for their future: her initial statement that she does not know whether their

¹⁶² It is surprising that Stacy views her son as having "always been really well" as he has needed to be hospitalised approximately twice a year for the last five or so years. Obviously, she gauges his health in relation to other children with the disease.

luck is “going to change” borders on denying that the disease will progress in her child (but surely she views her child’s life in parallel with the course of the disease?); her revised statement that “it probably will” acknowledges her (and my) knowledge of the nature of the illness but leaves open a chance that somehow her son’s fate is not pre-determined; finally, she admits it as inevitable that “he will get sick” but she pins no certainty to what she means by this. Even now it seems she uses the word ‘sick’ as a euphemism for what must come to be – as a reference to something which better remains vague than concrete – and in so doing, she keeps thoughts of the future in abeyance.

So much is she immersed in the ‘everyday’ that it seems she leaves even the ‘what-has-been’ behind. By Stacy’s own account, when life is going along smoothly and her son remains quite well for long periods of time she seems to forget that he has a life-threatening illness; she forgets the significance of his numerous hospitalisations over the years, the needles, the medications, the doctors’ appointments...

I think because he was so well for such a long time, and then when he is sick then it kind of like hits me more (2: 50-52) [...] I don't really think about Bradley being sick at all until he is sick, and then I think, "Oh shit, he is really sick". (3: 5-6)

This ‘forgetting’ teamed with a lack of ‘anticipating’ means that she lives insulated in the present, allowing her to be lulled into a sense of normalcy. While this allows her to cope and to function in the here-and-now when all appears well, it comes at the expense of having to endure a harsh reminder that he is not like other children each and every time she sees him get ‘sick’:

Like when he’s got a really bad cold and the doctor, and I go to the hospital and they say that he has to go in – two weeks I.V., and then it kind of like hits me again that he is sick, and I kind of like stick it in the back of your head all the time...that he is – has got something wrong with him, and then like when you go to the doctors and they tell you he has to go in, you think, “Oh shit, he really is sick”. Then it kind of like hits you again. But the first time he [...] had to go into hospital, he was about seven, and I think it was...[long think]...it hit

me like it did when I first found out that he had CF 'cos he had never been into hospital and all of a sudden they say, you know he's gotta go into hospital and I thought, "Oh my God, he is sick" (3: 10-18).

For Stacy, hospitalisations are the periods of time when she considers her son to be a sick child. This is the trigger that brings her son's illness to the foreground of her existence. When he is well, she allows it to fade into the background again for the time being.

Flynn states that the existentialist sees "the value and meaning of each temporal dimension of lived time [as] a function of our attitudes and choices" (2006, p.5). Accordingly, he says our temporal priorities are established based on our life decisions (2006, p.6). Stacy's temporal priorities are based in the here-and-now. Her experience living with her son's disease has taught her to be grateful for the gifts of the present and that energy spent worrying about the yet-to-come is wasted energy. Flynn refers to the adage, 'time is of the essence', as one that existentialists would insist can be interpreted in a very literal way for they contend that "part of who we are is our manner of living the 'already' and the 'not yet' of our existence, made concrete by how we handle our immersion in the everyday" (2006, p.6). If "[t]ime has its own viscosity" (Michel Foucault n.d. cited in Flynn, 2006, p.6) and lived time "embodies its flow" (Flynn, 2006, p.6), then Stacy can be seen to be fixed in a slow-moving molasses that usually stays put until a good dose of external energy causes it to flow in one direction (to the 'already') or the other (to the 'not-yet').

Stacy's lack of attention to the future extends to her parenting of Kristy. As the interview unfolded, it became clear that she has never thought ahead to the time when she will need to tell Kristy she is a CF carrier and how it will be best to do this. She did not have much to say on the topic without probing or specific questions. One thing she was adamant about and repeated over and over throughout the interview was that she herself would never have a child with CF again. And while she makes it quite clear that she recognises it would be a decision for Kristy alone to make, she hopes that she too would not go ahead with a CF-affected pregnancy:

[I]t's a lot of work, you know, kind of like your whole life changes. It's not like having a normal child at the beginning... 'cos you do, you wrap them up in cotton wool, you don't go out anywhere. Well, I did anyway. And then, I think if [Kristy] doesn't wanna have to go through all that – just wants to have a normal, simple life with a happy, healthy baby... (12: 9-13) [...] I don't think I would like to see her with a child with CF because of all the stuff she has to go through – I just want her to be happy and have a normal child. [pause] But if she chooses the path of having that... I mean I'll help her every way I can. It's just that for her... I don't know – going through what I went through, I think it's... [...] If I had to do it all again... I mean I would do it because I've got [hesitates] a beautiful boy out of it... but, I would not go through it again – like I wouldn't have another child with CF, I just wouldn't do it. (12: 19-25)

Her focus on “all the stuff she has to go through” seems to be largely about all the hard work that is involved in raising a child with CF in the early years, rather than about the emotions she may feel or the prospect of losing a child at a young age or whether there is a moral dimension to knowingly bringing a child with a disease like CF into the world. In saying, “going through what I went through”, Stacy relegates the impact that having a CF child has had on herself to the past rather than referring to it as a present and ongoing thing that she will have to continue to deal with in the future. Not once in the interview did she talk about or refer to what she will have to go through as her child inevitably gets sicker as the disease progresses. For me, I imagine this would be the hardest thing about having a child with a disease like CF – knowing what the future holds, anticipating the pain you will go through and feeling anguish before the realisation that your child will grow to become aware that they have a much shortened life expectancy, yet Stacy never refers to this aspect of the disease. I suppose it is probably too painful to think about. Is this a coping mechanism? Denial? Possibly both.

The advice she suggests she would offer to other people living with the knowledge that their child is a carrier and dealing with how and when they will tell them about it reveals the philosophy she tries to adhere to in her day-to-day parenting; it is best to focus on the positives of today rather than anticipatory concerns for the future:

*I would say: "Don't worry about it. Just thank God that your child doesn't have CF and deal with it when the time comes...[quiet laugh]...that's what I would say – don't even...I just figured, think that um...that you're lucky you've got a happy, healthy child and **why should you worry about something that's going to happen twenty years down the track?** You know, why put something like that in your head when you don't have to do it right now? That's what I think, with anything. Like even with Bradley, why worry about what's gonna happen in five years time, you know, just worry about what's going to happen tomorrow or next week, you know. That's what I think. (14: 52 – 15: 4)*

It struck me that this was the time she spoke most confidently in the entire interview. I think when I asked her about things she had not thought about before, she felt like she had to come up with the 'correct' opinion on what I had said when perhaps it did not have any applicability to the way she felt or the way she tends to think about things. Yet, when I asked her this question, she had an opportunity to have a 'final say' and to make it clear that this is not something that parents should worry about or invest too much energy contemplating over. Again, she compared the carrier state to having the disease itself and by doing this she can view it as a positive thing, relatively speaking. Having a child with a chronic illness seems to have made her more grateful for the gift of health, with the rest all being relegated to the category of 'small stuff'. She emphasises the most important thing is that "you've got a happy, healthy child".

I wonder if many 'CF parents' live in the here-and-now and, if so, if this is because the here-and-now can be dealt with and managed while future events may seem out of their control and therefore better to avoid thinking about. Existentialists attach extreme importance to the role that the future plays in explaining human behaviour – "a person's existence at any given time is incomplete or 'unsaturated', since the person is always 'on the way' to becoming something in the future" (Cooper, 1990, p.72). Cooper elaborates on this by explaining that my present situation comprises facts about myself which take on significance for me precisely in light of what I am 'on the way' towards (1990, p.73). He continues, and ends by paraphrasing Heidegger (1962):

This is part, at least, of what several existentialists have in mind when they insist on the logical priority of a person's future over his past and present. 'The primary meaning of existentiality is the future' [...] It is the future which 'first of all awakes the Present'. (Cooper, 1990, p.73)

If this is the case, one wonders if Stacy's way of existing – wholly immersed in the present – somehow limits her possibilities. If the future awakens the present, and if the life 'projects' I am on the way toward realising lend significance to aspects of my present situation, how can Stacy make sense of her present when she claims not to live towards a future as such? What happens when an individual's present assumes 'logical priority' over their future? How does this play out in the life of a person like Stacy? Are there greater ramifications for her beyond solely her orientation to parenting?

Waiting for the Right Time

MAKING A JUDGEMENT CALL ABOUT TIMING

Timing is important to the custodians of genetic information. They form judgements about when the right time to release that information is but the factors they base this on vary. Some plan for it to be based on contextual factors, where they would like the information to flow out of a more natural conversation; for others, it is an age/development-related decision; some look to the best way to minimise worry, where they believe timing will have an impact on this.

Vicki and Tim reported having "thought a lot" about when the best time will be to tell their seven-month-old son that he is a carrier (18: 10) but said they have not "actually come up with any solution" (18: 10-11). As Vicki points out, "[they have] got time obviously" to think about it (18: 11). However, of the three possible options they mentioned, it is clear that they will be looking for something to trigger 'the conversation' – awaiting a context within which it can naturally arise – rather than using their child's age for a gauge as to when he will be ready. The following are the options Vicki spoke of and her brief thoughts on each:

[1]] *If he starts doing biology class and starts to learn a little bit about genetics, then he would have a bit of a foundation for that knowledge. (18: 5-7)*

[2]] *[W]hether we wait until he meets somebody but then, you know, at what time of that relationship do you bring that up? And then he would have to make the choice whether he would tell his partner. (18: 7-9)*

[3]] *[I]f there was anything on television or whatever then that might be...pre-empt us to discuss it (19: 3-4).*

All in all, she agreed that it would end up being very much a wait-and-see kind of approach (19: 7-9).

Josie was one of the mothers who mentioned age and other development-related factors when assessing the appropriate time to discuss her nine-year-old child's carrier risk with him:

[H]e's got to have this specific test – it probably would be good if we¹⁶³ had it when he's late in his teens before he settled down and had children and whatever. (17: 15-17)

Though not explicitly expressed, her concern seems less about when he will be old enough to make a decision himself about testing than when he will be old enough to understand the genetics of it:

I mean I could, as I said, have [his carrier test] done tomorrow and tell him. It wouldn't really make much difference to me but he wouldn't understand it, tomorrow, you know like if we told him. I mean he probably would if we sat down with him and told him everything but yeah, just...you know like, he's not going to go and have children tomorrow so why worry about it, you know like? (17: 17-21)

We see that there is no room in her thinking for the idea that he may make the decision not to be tested at all, only that 'they' may delay testing until "whenever he really wants to – if he's not up to it then" (9: 53).

¹⁶³ Note Josie's use of the pronoun 'we' when discussing decision-making about her son's test.

On when she will tell her daughter about her carrier state, Tanya said it will be some time in her teenage years “and that’s years away yet so I haven’t thought too much about it really in-depth” (13: 19-20). It seemed Dawn too had not given it a lot of thought since moving beyond the time of it being “[p]anic stations” (9: 1) when there was a chance her daughter had the disease. This was revealed through conflicting data in her interview. At one point, she said

it’s something I’ll discuss with her as she gets older, [pause to think] especially more when she’s around the age of having kids or if she’s in any partnerships or things that looks like that she will have children. (8: 29-32)

Then only a little later she said, “I daresay it’ll be more discussed when she’s going through puberty” (9: 3-4). Her musings show that she does not have a fixed, set idea of when she will tell her daughter but that she does see it being linked to a point in her development.

Kath’s three youngest children are in their late teens and have not been carrier-tested. Kath does not think they know their nephew is a carrier because they were young when it was picked up via the neonate screen eleven years ago and it is not something that has been discussed recently (11: 47-53). Accordingly, they are unlikely to be aware of their carrier risk (11: 55-57). She admitted she is “not sure when or how that should be looked into” (3: 44) – when she approached the family doctor about it at the time, he simply said “Oh, there’s plenty of time for that when they’re older” (3: 46). Because of this, their carrier risk has not really played on her mind a lot (8: 51-54) but now they are older she would like them to be tested fairly soon – she “would like to know” (9: 16-17). She is a bit confused about where to go from here though:

[I]t’s a bit of a worry with the young ones to know how we go about it or what we should do or just ignore it until they are old enough to decide. I’m not really sure on that one. (5: 53-55) [...] [I]t’s no good badgering them saying you’ve got to have it done if they don’t want it done, or they don’t think the time’s right – that’s fair enough. But

if they are happy to have it done soon, I think it should be maybe done. The girls are too young for relationships. [My son] has got a girlfriend and...but...like that's what makes you think about it more, when they have got a girlfriend or boyfriend – more than when they're younger. (13: 35-43)

We see that, like Josie, Kath presumes her children will want the test but she too recognises that this will need to occur when they are ready to go down that path themselves. However, unlike the other mothers mentioned here, Kath does see some benefit to addressing it sooner rather than later:

If they get tested soon and none of them are carriers, it's not really something they need to tell anyone. [...] So that's why, I suppose, it's...rather than waiting till they are in a relationship and then spring it on someone, at least they can be prepared, and if [my oldest son] was the only that's got it then it doesn't really need to be worried about. (13: 49-56)

So, for Kath, while the 'right time' may include aiming to minimise the number of people who will need to **worry** about carrier test results, it also gives consideration to minimising the number of people who need to **know** about the family 'trait' – not surprising given she is one of the parents who felt there was some stigma attached to having a disease gene in the family (see section: Shame and Stigma Associated With Having 'Something Wrong').

There were other instances where information custodians revealed there may be a 'right time' to pass on genetic information in terms of minimising worry. In contrast to Kath's idea on this though, this often involved delaying it rather than bringing it forward (for one example, see section: Protected Rights of the Child). Leanne and Rob certainly felt this way in terms of genetic information that was relevant to his sister which they had not yet passed on to her. Apparently she "has talked about having children in the next two years" but despite a friend of Leanne's advising her that it may be better "sooner than later" to tell her of her risk (9: 28-31), they still have not done so. Rob intimated that he had held off doing so, initially because they thought she was not planning to have children (see section: Leanne's Call: "There's No Point Telling Them If They're Not Having Kids"), but, later, because he

saw no point in “letting her worry about it two years before she’s even ready to [have children]”. However, Rob began to rethink this as it occurred to him that having more time may help her adjust to the information:

Rob: *Although, you know, maybe by then that [extra] time would’ve helped. I mean, it’s probably helped us cope with finding out about the whole thing. As time goes by, it becomes easier to understand and live with.*

Interviewer: *So do you think in some ways you are sort of protecting her a little bit?*

Rob: *Probably.*

Interviewer: *Mm. From...mainly from the worry of the possible consequences?*

Rob: *Yeah.*

Leanne: *Yeah. I think that’s fair. I think if you don’t...yeah if, you know, if she’s gonna worry about it for ages, I think that doesn’t help at all but in the same sort of sense you do need to give her enough time beforehand to sort of just digest it and realise that, you know, it’s only a chance thing and they can go, and like you said, you know, go and have the test done and find out that they’re not both carriers. Well then, that puts all their worries at rest and then it’s done. So that’s probably something that we should do soon. (11: 2-21)*

We see that for Rob and Leanne it is a fine line between imparting the information too soon and leaving it later than is helpful. The danger is that in their concern to protect his sister, they may leave it too late – what if she and her partner wish to surprise the family with an earlier-than-expected pregnancy announcement? We see that their focus on ‘waiting for the right time’ has the potential to restrict another stakeholder’s reproductive possibilities.

Timing can be everything.

OUR CONTINGENCY¹⁶⁴

Being Lucky

THE CHILDREN WHO ARE 'ONLY' CARRIERS

Parents often expressed relief and felt their children were lucky – even blessed – to be 'only' carriers.

Stacy reported that, in revealing to her daughter that she is a carrier, she plans to emphasise to her daughter how lucky she is not to have the disease (7: 26-27), that "it's better to be a carrier than it is to have cystic fibrosis" (8: 4-5) and she

*just hope[s] that [Kristy] takes it as **she's blessed that she's only a carrier** and not that she's going to be so upset that she's not ever going to have any children or anything. (8: 29-30)*

Don and Betty had two healthy, much-older daughters before the arrival of their third child, Elise, who is affected with CF. Despite both the older daughters being carriers, like Stacy, Betty focuses on the fact that they are healthy: "[A]ren't we lucky? We got away with it twice [before Elise came along]" (15: 33). Adding to their 'luck', she comments on the realisation that had either of the two much older daughters been born with CF, they would have been in the generation with a poorer prognosis than that of Elise's (15: 34-36): "A lot of those kids are dead so...we're very lucky" (15: 40). Betty looks to the positive aspects of their situation, creating a narrative that comforts them in the knowledge that things could have been worse.

Betty said that neither she nor Don were "overly [worried]" when they found out their girls were carriers because they knew this would not have any impact on their health (16: 51-54):

We just thought "Oh, bad luck", you know, we could've had children who were clear of it but we didn't. We're carriers and it hasn't really affected our life until the two of us got together. (16: 54-56)

¹⁶⁴ The contingent nature of our existence was discussed in Chapter Three (see section: Nausea).

And as for how their girls themselves feel about being carriers, Betty said that they have

had it instilled in them that Elise was the one that drew the short straw and they're the lucky ones so I guess from that point of view, they've always thought they've been the lucky ones. (23: 21-23)

Betty, like Stacy, reveals that she has taken it upon herself to try to shape her daughters' perceptions of their own carrier status, drawing to their attention how much better off they are than their sibling with the disease. While this concern obviously stems from parental love and a desire to safeguard her daughters' emotional health, it is possible that reinforcing the idea that these children are lucky in relation to their sick siblings could augment their experience of 'survivor guilt'¹⁶⁵.

Following an initial period of panic when told the health of their newborn babies was under a cloud following a high-risk result for CF in the Guthrie test, parents from families where there are no instances of the disease also spoke in varying degrees of their eventual relief in discovering their newborn babies were only carriers. As simply stated by Tanya, "[W]e were just glad that she didn't actually have CF" (9: 27).

More than merely being glad that her daughter did not have CF, Leanne provided an animated, even euphoric, account of her reaction when told that her daughter was *only* a carrier:

*It was really just that relief that "Oh gee, she's only a carrier – that's fantastic". That's like...**that's like the best news in the world when you find out "OH YAY! She's a carrier!"** [sounding ecstatic] It's like almost exciting. It's like "Yay!" because, you know, that's so much better than the other option so... (12: 56 – 13: 2) [...] [It was] just a*

¹⁶⁵ 'Survivor guilt' is a phenomenon that has been described in relation to favourable genetic test results previously. In one quantitative study which examined the psychosocial impact of presymptomatic DNA testing for Huntington's disease, most of the newly-identified carriers seemed to function well but a surprising result was that in eight out of the nine non-carriers, "the expected relief was short-lived and was soon replaced by persistent guilt feelings (survivor's guilt), depression, and emotional numbness [...] All non-carriers experienced feelings of guilt towards their affected or at-risk relatives. They felt an obligation to be continuously available to bolster affected or at-risk members of the family" (Tibben et al., 1992, p.97-98).

positive thing that she was just a carrier so it wasn't definitely...I don't see it as a burden... (17: 31-32)

Vicki spoke of her and Tim's relief (3: 25-26), describing the moment when the news was first conveyed to them (a voice message left on their phone by the registrar) as one of joy:

I think we both felt like having a bit of a dance [laughs] [...] And [our baby boy] got more hugs and kisses! (11: 40-41)

Until that moment, since being informed of their son's 'high-risk for CF' status, Tim explained that he and Vicki found themselves in the curious situation of 'hoping' their son would be a CF carrier:

[W]e just hoped and prayed that there wouldn't be anything wrong with him and that he'd just end up being a carrier – although it seems like a weird thing to hope your kid to have. Just hoped that that would be what it was. (22: 50-53)

The couple do not regret the fact that they have been given the information that their son is a carrier of cystic fibrosis, always juxtaposing talk of his one CF mutation with the at-one-time very real possibility that he may have had two:

[W]e see [the neonatal screening program] as a positive. Like, we don't see the whole thing as like a... We always try and look on the positive side of things and, you know, we are very lucky that Zane doesn't actually have cystic fibrosis. We don't look at it, 'Oh my God, he's got the CF gene'. It's, you know, the focus is that he's a healthy boy. (Vicki, 29: 4-7)

Overall, most of the parents perceived their children's carrier state in a positive light because they were comparing it to the alternative scenario of being affected by the disease itself. While this was probably predictable in families where the child had a sibling or a cousin with CF, all the families with a child identified as a CF carrier by the neonate screen had been through the trauma of being recalled to the hospital and witnessing their child undergo a sweat test while they awaited a potential diagnosis of cystic fibrosis. Hence, despite the lack of prior history of CF in these

families, their anxiety about the disease had been heightened and it was a great relief for them too to find out that their children were **only** carriers.

In interpreting this being 'only' a carrier as a 'blessing', the parents view existence in a manner akin to Gabriel Marcel – as “a gift to be accepted in a spirit of thankfulness” – rather than, as Sartre would say, “a brute fact to be dealt with” (Flynn, 2006, p.60). These parents' relief speaks of an all-too-keen awareness of the arbitrariness of procreative genetics and the resultant gratitude that comes with recognising the good fortune that befell their healthy, albeit carrier, children.

A LIFE OF SHEER 'LUCK'

While the two mothers who had grieved the loss of child/teenage siblings to CF certainly acknowledged they were extremely lucky not to have been afflicted with the disease themselves, they did not necessarily see this in a positive light. Neither expressed anything like relief or joy and one spoke of the guilt she felt as a child that it was her brother alone who was affected.

It could be said that the two adult CF carriers in this study who have lost siblings to the disease have had to face their own contingency more intently than most because they realise their good health and continuing existence into adulthood have been the result of completely random, once-off meiotic¹⁶⁶ events – that it could have so readily been them instead of, or as well as, their deceased sibling(s), who were born with the disease.

The characteristically existentialist issue of our contingent existence is “born of the experience that we are, as it were, forlorn castaways”, or as the musician, Bob Dylan, put it, “I was born here and I'll die here against my will” (Marino, 2004, p.xiv). This lyric leaves us unable to escape acknowledging the whimsical nature of the way

¹⁶⁶ Meiosis is the name given to the nuclear divisions in the special cells in the gonads that are destined to produce gametes (e.g., sperm and ova). The result of each meiotic event is that one cell with two equivalent chromosome sets is divided into four meiotic products, each with only one set of chromosomes.

we came into existence and one would think this must be heightened for the healthy carrier siblings who just escaped “the double-barrel; the CF” (Don, 15: 30).

Wendy has one copy of the CF gene mutation and lost her younger sister and brother to CF less than a decade ago. As she reflects on her childhood, it is clear that her family doctor was influential in impressing upon her the contingency of her own existence, even as a child. While she acknowledges the family doctor’s conduct towards her reinforced how lucky she was to have escaped the disease, she said it did not make her feel any better about it:

Wendy: *I always remember them telling my Mum that she shouldn’t have any more because the chances – she’s had the one – the chances are she’ll more than likely have another one. Just sheer genetical luck. It’s got it written on one of my files somewhere...yeah, “Sheer genetical luck”, my doctor wrote [difficult to hear this because she cried/laughed suddenly as she said this last bit – very upset].*

Interviewer: *Oh, about you?*

Wendy: *Yeah, well I suppose it is ‘cos if I’m a carrier and I haven’t got a disease and both my siblings have...whooooo [makes a ‘that was close’ sound] – just a cell divided in the right spot, didn’t it? I suppose. That’s what it all boils down to, isn’t it?*

Interviewer: *Well, I guess, I mean, in terms of the genes that you got from your parents – it is luck. But you know that actually a carrier has like one copy of the CF gene, and people with CF (the disease itself) have two copies of the gene.*

Wendy: *That’s right, yeah. That’s why...yeah, it was explained to me because I’ve got one good gene and one bad gene and that’s why I’ve got good genes and they threw in a bad one! [laughs at herself] So yeah, I understand all that.*

Interviewer: *So when you say the doctor wrote on your file, “Sheer genetical luck”...*

Wendy: *Oh, that was years ago. Yeah, that was years ago. Yeah, he used to sit there and just look at me – every time I went to the*

doctor's for a check-up, which wasn't very often – he just used to sit back and I remember him sitting and just shaking his head [does the motion as if to say, "It defies explanation that you're healthy"]. [laughs heartily] Just shaking his head.

[...]

Interviewer: *So how did that make you feel? Do you remember how that made you feel?*

Wendy: *Oh, it never used to make me feel any better, I suppose.*

Interviewer: *It didn't?*

Wendy: *Oh, I don't know, just lucky I s'pose. It used to make me feel lucky. I learnt to appreciate life at a very young age anyway, I know that. I learnt very young that everything in this world is material – it means nothing really. Can't take it with you if you die. [silence] (2: 37 – 3: 27)*

Wendy hones in on and perfectly articulates the crux of the matter of her contingent existence when she refers to “just a cell [dividing] in the right spot” as being all that her health and continuing existence boil down to. It is noteworthy, however, that an acknowledgement of luck around the genetic cards she was dealt does not mean that this actually makes her feel “any better” about escaping the same fate as her siblings.

Cindy also described feeling lucky that she “missed out” (7: 54) on the disease (her younger brother died due to CF almost thirty years prior to the interview). In response to a question about how she feels that she too could have been born with the disease:

Lucky. Lucky. And when I was younger, I did think about that. But I also thought the whole three of us could have been born with cystic fibrosis or the whole three of us could have been any combination of... So yeah, and I guess that was...when I was saying before, “As a child, I felt guilty about his death”, it was kind of linked to that too because it could've been me but it wasn't. And why wasn't it? And

what did that kind of mean? So you do...I mean you don't have one isolated thought – it all interrelates so... (8: 8-13)

Cindy spent time ruminating over the genetic roll of the dice in her family too. And for Cindy, as with Wendy, her luck did not bring relief – only confusion and guilt about her own existence; ‘survivor guilt’.

Fixating intently upon one’s own non-necessity can be a disturbing and troubling experience, as articulated by the central character in Sartre’s *Nausea*:

And I – weak, languid, obscene, digesting, tossing about dismal thoughts – I too was *superfluous*. Fortunately I didn’t feel this, above all I didn’t understand it, but I was uneasy because I was afraid of feeling it (even now I’m afraid of that – I’m afraid that it might take me by the back of my head and lift me up like a ground-swell). I dreamed vaguely of killing myself, to destroy at least one of these superfluous existences. But my death itself would have been superfluous. Superfluous, my corpse, my blood on these pebbles, between these plants, in the depths of this charming park. And the decomposed flesh would have been superfluous in the earth which would have received it, and my bones, finally, cleaned, stripped, neat and clean as teeth, would also have been superfluous; I was superfluous for all time. (1963, p.184-185)

While the character is logically aware of his superfluity, he expresses fear before the thought that this awareness may turn into a wave of emotion that overwhelms him.

People who seek solace in religion are able to take an alternative, less futile view of the superfluity of their existence as individuals. .Says Flynn:

Theists [look] to a Deity that understands and cares. For them, freedom is genuine but created. **They view the world and our existence as a gift** and an invitation to a loving response. Our resultant attitude should be one of what Gabriel Marcel calls ‘creative fidelity’ to this gift. [emphasis added] (2006, p.55)

While Cindy’s views on religion were not discussed, she speaks of her own children’s good health and both her parents’ and her ex-husband’s family’s wellness

as if these may be 'gifts' of a kind to compensate for the suffering her family has had to endure:

*[A]part from [my son having an operation for double inguinal hernias], the kids have had relatively healthy lives. [My daughter] gets a bit of tonsillitis every now and then but that's it. [chirps up a bit] **Maybe in a way I think that's a pay-off.** [nervous laugh as she thinks about the irony of having two very healthy children when they were at risk of being so sick] You know... [trails off as she thinks] Both my parents are in their 60s and well, and both their families have been well, and my husband's family are pretty well. [...] [By 'pay-off', I mean] **if you don't get [CF] then you're well** [said bluntly]. You know, **like if you don't have the disease then like you've really missed out** (I mean, that's a silly way of looking at it). [...] **[I]f you don't actually have the disease, you're actually really okay.** You know like... [...] quite healthy. (15: 7-27)*

If these are 'gifts' then they are something to be grateful for, and this implies there is something or someone to be grateful to. Here again the concept of a paid debt in life emerges (as we saw with Josie; see section: When Termination Is Not An Option). In her own mind, Cindy has constructed the health and wellness of her family as a 'pay-off' for the tragedy they have had to suffer through. Her words betray an underlying need to believe that there must be some overarching schema in place that ensures some degree of 'fairness' to human existence, despite her early life experience so very much to the contrary.

Of note here also is the fact that Cindy categorises family members only as **either** having the disease **or** being healthy, rather than delineating a third interim state for those who have one copy of a CF gene mutation. This is consistent with the interpretation of data from elsewhere in her interview that the carrier 'state' is one of **knowing**, not **being** (see section: The Carrier 'State' Is One of Knowing, not Being).

OUR FACTICITY AND OUR TRANSCENDENCE¹⁶⁷

Sartre is [...] intent on underscoring our responsibility for the necessarily ambiguous situation in which we live. Whatever our situation, it always includes the possibility of moving beyond it. [...] [T]he mantra of Sartrean humanism is that you can always make something out of what you've been made into because you always transcend your facticity. (Flynn, 2006, p.67)

Passing on the Gene

THE GENERATION GAP

It seemed the case that grandparents of a child with CF generally dealt less well than parents with the knowledge that they had passed on one of the genes that caused the disease.

For the parents of a child with CF, their focus was certainly on the task at hand – coping with caring for their child and trying to keep them well:

Yeah, we were upset I suppose in a way – that we'd passed it on to this innocent child sort-of-thing and now she's got to live with it. But it was too late, you know, we couldn't do much about it so we just had to not dwell on that, like look forward to what we've got to do sort-of-thing. (Josie, 8: 15-18)

Perhaps having a job to do, with tangible outcomes, gave them some control and a sense of power over a situation that was otherwise beyond their control and over which they were powerless – the birth of a child with a genetic illness, previously unheard of in their families. Josie said she had not focussed on her own carrier state much. She explained that her main concern was looking after her affected daughter and bearing in mind that she and her partner would need to “know the risks” (7: 54) when having more children in the future. But, she reported that her own mother had been quite upset to find out she was the carrier; that she had

¹⁶⁷ The duality of the human condition as both facticity and transcendence was discussed in Chapter Three (see section: Ambiguity).

passed this gene on to her grandchild. Josie said she herself was the one who tried to make her feel better:

I said, "Well, it's not just you". [...] I sort of had to explain to her, "Don't worry about it". You know like she seemed more upset than I did sort-of-thing about it. Because, I mean, my Mum's quite elderly as well so she couldn't really get her head around it too much as well. But she sort of blamed herself a lot and I said, "There's nothing you can do. It's in your genes – it's not something...it's been passed on to you." She was quite upset about it. (8: 10-15)

She contrasted this with her partner's father who, while being a bit upset about passing the gene on, was able to rationalise it to himself thus:

[H]e wasn't too fussed 'cos he sort of understood "Well, it's been passed on to me and now I've passed it on but we never knew nothing about it" so... (8: 19-21)

He accepts the facticity of his situation – his powerlessness over what genes he passed on to his children.

Henry, a carrier, and grandfather to a child with CF, sounded sad and quiet at times when talking about passing the CF gene mutation on, yet he expressed resignation about it and seemed to have accepted that it was something over which he had had no control:

But what can you do? I mean, you can't change it now any more. (2: 22) [...] [I]n one way, I just live a life and, yep [sounds wistful, voice a bit shakier for a moment]...it is terrible what can happen to a child, you know... (2: 32-33)

His expressed hope was that not too many others in the family will have this same gene (2: 22-24).

Don and Betty both got the CF mutations they passed on to their daughter from their fathers. Betty said her father

felt a little bit almost responsible initially, just momentarily [...] my Dad felt a bit...a twinge of guilt that he had passed it on to me and

passed it on to Elise and then we had CF. But I think everybody soon realised that we've got no control over that – it's just part and parcel really. [Long pause] As far as us being carriers, like to me it's nothing compared to what she's got to put up with. (15: 22-27)

Don said both he and Betty accept that they have passed on the gene that is the cause of their child's disease, and he "[does] not feel guilt or anything like that" (15: 29) but he does question why Elise "had to be the one to actually get the double-barrel; the CF" (15: 30). In contrast, Stacy, also a parent to a child with CF, said she does "feel a bit guilty being a carrier [...] and then having [my son] and giving it to him" (22: 12-13).

Betty believes that a lot of older people feel guilt about passing on the gene because they do not properly understand the genetics of it but she admits that if one of her daughters who carries the gene has an affected child, she will

probably still feel a twinge of "Oh gee, it's come from us", you know. Yeah, I guess so, as a grandparent. As a parent, [...] you're just that step away and you don't like to think of your children suffering. But your grandchildren...you don't want to see your children and your grandchildren suffering! With Elise, we've got sort of complete control over her, but grandchildren they've got to cope with it in another environment, another house...and we know the stress it's caused in our house and I'd feel guilty – I'd think "Oh gee, I've burdened my children with that now!" (17: 51 - 18: 1)

There seems to be a generational difference in the emotional response to passing on the gene, namely that the grandparents seem to feel more of an ongoing sense of guilt and blame than the parents do. It could be that in their role as primary carers of their children, the parents have the opportunity to reclaim power (keep their children well) amidst their powerlessness (having had no control over passing on the disease gene to their children) but the grandparents do not have the opportunity to redeem their powerlessness in such an active way – they do not have access to that equivalent level of power.

The hypothetical guilt of which Betty spoke is one that Rose's Mum expressed relief to be personally freed from. As the grandparent of a little girl with CF, she said it was a relief to discover that she does not carry the familial CF mutation and that, therefore, it must have been her ex-husband who passed on the faulty copy of the gene to their children. She explains that

as a parent, or as a mother...you, anything your child gets, you automatically assume that it's your fault – something you've done or something you haven't done or what-have-you...and to have a huge thing like that, and then to find out that you couldn't have prevented it anyway, it is...it is a relief because then you can put your energies into not feeling sorry for yourself... Because I mean you do, you do, you really feel sorry for yourself 'cos you think you've inflicted this on them but then you can throw all your energies into helping them overcome their fears or whatever they feel. (3: 55 - 4: 5)

Rose's Mum's assumption of being at-fault when her child has emerged into the world less-than-perfect harks back to the 'maternal imagination' hypothesis, which for centuries has been used to explain the 'monstrous birth' (Braidotti, 1996). In teratology – the science of monsters about which Rosi Braidotti has written – the term 'monster' is used to refer to the deformed or anomalous body¹⁶⁸. In the sixteenth century, Ambroise Paré described the monstrous birth as "a sinister sign ('*mauvais augure*') that expresses the guilt or sin of the parents"¹⁶⁹ (Braidotti, 1996, p.139). The theory of the maternal imagination "attributes to the mother the capacity to undo the living capital she is carrying in her womb; the power of her imagination is such that she can actually kill or deform her creation" (Braidotti, 1996, p.145). As new genetic technologies allow us to see more imperfections at the molecular level, a new range of biological 'deformities' is emerging. And while the gene carrier is a monstrous form far beyond what Paré could himself have

¹⁶⁸ Braidotti says that "[t]he quest for the *origin* of monstrous bodies has motivated some of the wildest theories about them" (1996, p.139) and explains that use of the word in this manner evolved from the Latin term, '*monster*'/ '*monstrum*', which is primarily an object of display (1996, p.135). 'Monster' is a label that was applied to the unfortunate human 'exhibits' in royal courts, 'freak shows' and circuses over the centuries.

¹⁶⁹ In Paré's day, it was thought most likely to point to an unacceptable sexual practice having been indulged in on the part of the parents during the act of conception.

imagined, it has the same existential import as those of which he spoke. Rose's Mum's statement here attests to this.

Another grandmother, Kath, talked about the helplessness she would potentially have felt if one of her children had a child with CF and had to bear the burden 'alone' (without her help). It was because of this that she was "relieved" (8: 29) her two oldest girls, in particular, tested negative for the familial CF mutation:

Especially, it was a bit harder because they were both living interstate – both children are interstate – so if they had have had a child with cystic fibrosis or they were a carrier and their partner, it would have been a lot harder for me to accept being so far away and not being able to help them. (8: 29-33)

By her own account then, if Kath's daughters were carriers, knowing she had the power to 'help' should the worst happen (her transcendence) would have assisted Kath in accepting her powerlessness over which of her genes had been passed on to her daughters (her facticity).

CARRYING ON THE LEGACY

In two of the interviews the notion of one person alone being responsible for leaving an undesirable genetic legacy to their descendants was touched upon. Those who felt this seemed to see themselves as the sole conduit for the gene, without whom the gene would otherwise have been stopped in its tracks.

Wendy had been quite upset finding out she was a carrier initially (even though the carrier test for her partner at the time was negative); she "cried for a while" (13: 56). It "just made [her] worse" (14: 1) when her grandmother asked why she was so upset about it when she had always known she probably did carry a copy of the faulty gene (indeed, Wendy herself said, "I wasn't shocked...that I was [a carrier] at all. I would've been like shocked if I wasn't" (14: 28-29)). She tries to explain her emotion at the time:

[very teary - she speaks very slowly] To find out that you carry a gene that's like a shotgun bullet to someone's brain from the minute they're born, it's like... [trails off] I don't know... [long pause] (14: 2-4)

She goes on to speak of the gene as having the potential to carry the legacy from the spectre of her past forward, and of herself as a vehicle through which the horror may continue:

I was just hoping of some hope that I could put that childhood nasty disease behind me – that it would stop at me [very upset]. [...] I'd really like for it...I just want it to stop. Stop it dead in its tracks here... [pause] but fingers crossed, it did stop at me and it won't go on in my children. (14: 29-33)

This is quite unlikely given that, from a probability perspective, there is only a one in sixteen chance that none of her four living children will be carriers. Later in the interview, she acknowledges the likelihood that the gene's presence in her family is assured for at least another generation:

I was so hoping...I was so hoping that: "That's it – stop dead with [my sister and brother]. That's it. No more." Genetically, it hasn't really, has it...yet? (18: 11-13)

Wendy does not speak of a faith that Technology will ensure the healthy future of her descendants as many of the other parents did; her powerlessness over her situation here is evident.

This notion that one individual passing on a CF gene mutation is responsible for spawning an ongoing legacy for future generations in the family also arose in the interview with Anne and Kevin. After Kevin's brother had a child with CF, Kevin just accepted that it may be the case that he was a carrier too – he didn't even really ask questions about it until later (3: 14-15). He compared this to his own father's reaction to finding out he was a carrier:

I think it was much harder for our father to accept that his family had a history of cystic fibrosis. It explained a lot of child pneumonia deaths in the family going back. [...] I think it was harder for him to

accept the fact that that was where it was – that it came down that line. (3: 19-26)

Kevin's wife, Anne, proffered her opinion that it was just a matter of his family taking some time to adjust:

I just think it was adjustments to a frighteningly healthy [...] country family and they'd just never come across anything serious like that before. (4: 16-18)

But Kevin supposed that it had something more to do with his father's age and the era he grew up in:

[P]articularly for people who were brought up in the era that our parents were [...] went through the hard times and that sort of stuff and there was a fairly solid view that they had a responsibility to leave a positive legacy for their children, you know. And to get to a late stage, or a relatively late stage in your life – he was in his mid-fifties at that point – and find out that the legacy that you've left your children is actually potentially diseased children, I think it was a bit devastating, you know. (7: 13-20)

However, Kevin said his father is now "over that" (7: 24) and Anne attributes his coming to terms with it to knowing that

as [his grandchild with CF] has come along and, you know, [searches for the right expression] he's grown and he's a fully active member of the family and the community and everything else, I think that that initial rush of "Oh my goodness, what is this?" [...has dissipated.] (7: 29-32)

Anne believes this has allowed him to regain perspective and realise that

in the overall effect, he's actually been very successful with all his children and all his grandchildren (7: 27-29) [...] [I]n fact, he has six children who are very successful and very happy. (7: 26-27)

Curiously, Anne seemingly attributes the overall success and happiness of Kevin and his five siblings to their father and in deliberately using this as a counterpoint to the genetic contribution he made to his offspring unwittingly validates his feelings of failure on that aspect of his parenthood.

This aspect of the data is a departure from the findings reported in the study that looked at the processes employed within genetic counselling consultations (Armstrong et al., 1998). The authors found that a sense of an “individualised responsibility or culpability” for the threatened health of their descendants was *not* apparent as clients engaged with the newly-discovered knowledge that they carried a genetic ‘defect’ (1998, p.1656). However, that study was based on transcripts of “new genetic counselling consultations” whereas this study relied on data collected months or years post-counselling. Could it be that in some cases “the genetic nexus of relationships in which [a person is] embedded” – brought to the foreground within the confines of the consult as the search for an “origin” of the gene ensues (Armstrong et al., 1998, p.1656) – slips away as time passes and the person again becomes isolated in their individuality; focussed on their own personal legacy to their children, grandchildren and beyond?

(RE)GAINING CONTROL THROUGH ACCEPTANCE

Like most parents of a child with the disease, those carrier parents with only healthy children also expressed an acceptance that they could do nothing about passing the gene on and now looked ahead to what they could control; to where they could have some power over their situation.

Rose said that she does not feel responsible for passing the CF gene mutation on to her son but, it would seem, only because she anticipates that knowing he has the gene will mean he never has a child with the disease. She speaks as though the **onus of responsibility** is on her to ensure this does not happen, despite eschewing responsibility for his carrier state:

I don't feel as though like I'm responsible, or anything like that – I don't. I would have felt bad if he'd had a child in the future and I could have prevented it by knowing that I had it. I probably would feel responsible and feel probably a bit upset then. But no, I know that I have the gene – there's nothing I can do about it. But I know he has the gene but I know I can do something about it, by educating him now. (30: 14-18)

As discussed previously, in Rose's efforts to regain control over her genetic situation, we fear her 'education' of her son could jeopardise his right to an open future.

Wendy also expressed a strong sense of control amidst the uncontrollable when describing the prospect that she could one day find out that any one of her four children is a CF carrier:

I'll probably cry. There's nothing I can do about it. You can't change it. [...] I can't change the fact that they've got that gene but I can change the destiny of my grandchildren – that much I can do. (25: 45-51)

It seems that Wendy has taken it upon herself to ensure that her children transcend the facticity of their situation. And for her, this is the means by which she seeks to transcend hers.

Having eventually accepted that she had been powerless over passing the CF gene mutation on to her son and grandson, Kath also spoke of focussing on the practical courses of action available to her:

*I think at the time I didn't like that I'd done that but there's nothing that I can do to prevent it [...] 'Cos I didn't know I was a carrier so... (10: 29-34) [...] It's something that's in your make-up and you can't do nothing about it because it's been there obviously since birth but it's just a shock to your system and **once you do know, you want to find out more or do what you can to help the rest of your family or whoever.** (16: 27-31) [...] **Once you know, you know – and I think you're better knowing than not knowing because you are aware of it and can tell people or ring people or know for the rest of your family that it could happen.** (17: 18-20)*

Her acceptance of the situation despite her strong initial feelings are mirrored by Wendy's description of how she believes she will react over time if any of her children are identified as carriers:

*I'd just be devastated. It wouldn't worry me too much afterwards.
Couldn't change it. No matter what you do, you can't change it. (18:
6-7)*

Like Camus' Sisyphus, **resignation** to one's Fate – or at least, to the facticity of one's situation – brings **acceptance** that there are things in our life we cannot control. However, the paradox here is that these mothers believe control is possible through their grandchildren.

(RE)GAINING CONTROL THROUGH REVOLT

One mother reacted to her perception that the hospital staff were not taking her concerns and fears seriously by insisting that her baby son be carrier-tested. In an act of *Revolt*, she fought to secure a position as much like certainty as possible rather than accept the uncertainty of the genetic situation in which she was anxious she had placed her son.

Rose has a niece with CF. She reports having “to fight and argue to get [her first baby] tested” for the disease (sweat test) **and** the carrier state after his birth in the United Kingdom (4: 47-50). She claimed they did not have a neonate screen for CF there and the doctors questioned the need for it given that only she had been identified as a carrier (the baby's father had not been tested and so remained at the normal population risk). She was relieved to find out for certain that her son was a carrier because it meant he did not have the disease and it also meant her persistence in pushing for him to be carrier-tested as a baby – seemingly a symptom of her need for her concerns about the disease risk to be adequately listened to and responded to – had brought about a definitive result:

[I felt] relieved that I knew...now. Relieved that: 1) he wasn't...didn't have CF, which by that stage we guessed that he wasn't...but I was relieved to find out that – I finally found out if he was a carrier or not. 'Cos I felt I really needed to know that...for the future...I know I could've waited, I suppose...but the point was I got annoyed by that stage that nothing had been done from day one, when I first asked about [the disease risk]. (8: 16-20)

Rose's perspective on her interaction with these doctors was that she had to **battle** to be heard and she admits that in the end she persevered in insisting her son be carrier-tested because it had become a **fight** she was determined to win. It seemed important to Rose to confront the risk head-on and tackle it as soon as practicable – to eliminate **possibilities** by replacing them with **certainties**. Any anxiety she felt around her son's carrier risk seemed heightened by the idea that she should sit back and take a 'wait-and-see' approach as the doctors seemed to be advocating. And so, rather than resign herself to accepting all that could not be changed about her son's genetic situation and her biological part in it, she chose to turn the 'it was' into the 'thus I willed it' – attempting to transcend the 'givens' of her situation in an act of what could be described as *Existential Revolt* (Flynn, 2006, p.48).

Rose's Mum suggests where this relief her daughter speaks about comes from by comparing Rose's situation of parenting a carrier to her own situation of having a son (Rose's brother) whose daughter, Susie, has the disease:

*[As a mother,] you want to take these things away. You want to make everything right for your child – no matter how old the child is – you want to fix it all up and you know you can't [she is referring to her inability to lessen the pain of having a child with CF for her son]...this is something you can't...you can't, and it's something that [he and his partner] – you have to live with it too – but it's something that they really have to live with in their whole...in their everyday life. Every single day, it is there, you know, when you have a child with it. So consequently then when you are a carrier, I should imagine you feel the same...particularly like say Rose looking at [her two boys]...yeah, [her youngest son] could be a carrier – we don't really know at this point. But **you have such a relief that "yeah, so he's a carrier – we can do something about that" but with Susie we can't do anything.** (4: 10-20)*

Essentially, she is saying that parents make the best of a situation over which they fundamentally have no control by focussing on the power and control that a definitive carrier test result does give them. And **seeking control** is a typical response to the contingent nature of our existence as human beings. She explains further:

[W]hen you have a baby – when that baby is born – you want to do everything you possibly can to make sure that baby is fit and healthy and has a long good life, and if testing is going to help along the way, well yeah...you go ahead and do it. (23: 1-3)

She later added:

*I think there's a lot more to be said for the unknown than the known.
If you know what you're up against, you know how to deal with it.
But if you don't know... (30: 24-25)*

And yet, while all this explains why they think it is imperative that they find out the carrier status of Rose's boys, it is not adequate justification for pushing to have them tested at an age prior to them being able to have any input into the decision whatsoever. The right-in-trust of these boys not to seek definitive information relating to their carrier risk or to keep such information private (even from their mother and grandmother) should they decide to seek it out later has not been safeguarded by the genetic service provider of whom they too were clients with ethical demands.

CONCLUDING REMARKS

At a societal level we see about us a relentless drive to seek technological transcendence over our genetic situation. What troubles us so about the facticity of the genetic aspect of our existence? I believe Rosi Braidotti would answer: it is that we are afeared of the 'monster'¹⁷⁰ and what the birth of the monster signifies to us. In her analysis of the intersection between teratology¹⁷¹ and our perception of 'embodied differences', Braidotti summarises Ambroise Paré's examination of the 'monster' and the "illustrative function [it has served in] pointing out the infinite powers of the imagination":

¹⁷⁰ Braidotti's concept of the 'monster' was introduced earlier in this chapter (see section: The Generation Gap).

¹⁷¹ Braidotti defines 'teratology' as "the science of monsters" (1996, p.135). Its more common contemporary use is in reference to the study of biological deformities.

The kind of rarity represented by monsters is for Paré testimony to the ingenuity and the great variety of nature. Monsters are, therefore, not outside the natural order, but very much part of it, though they tend to represent the more fanciful and uncontrollable elements of natural life. (Braidotti, 1996, p.137)

Stigma theory tells us that we believe the monster – the deeply-stigmatised body – is “not quite human”, and this serves to justify our treatment of the monster as inferior and as representing a danger to us (Goffman, 1968, p.15). Monsters play on our fears about living a life that is ultimately beyond our control; that may be “changed from without” (Sartre, 1958, p.29). They provoke our *Sartrean Fear*¹⁷². Through their birth they remind us that we cannot dictate the terms of every aspect of our life, particularly not the offspring we produce. Monsters serve to remind us of our own imperfections and so we consider them as Other in order to distance the monsters’ imperfections from ourselves. Says Braidotti:

Discourses about monsters are fundamentally ‘epistemophilic’, in that they express and explore a deep-seated curiosity about the origins of the deformed or anomalous body. Historically, the question that was asked about monsters was: ‘How could such a thing happen? Who has done this?’. (1996, p.138-139)

Whereas we take great pride in our children being beautiful or strong or clever and tend to feel a sense of ownership or ‘craftship’ about the origins of these traits in them, we look to an outer source or reason for the monstrous birth, unable to accept the arbitrariness of biological development and once again our lack of control over the workings of our own bodies (*Sartrean Fear*). We need to remove the origin of this monstrosity from ourselves. We ask what we can do to prevent these births or prevent these lives which make our own so uncomfortable. Feelings of *Sartrean Anguish*¹⁷³ tell us that **there must be something we can do** to stop this discomfort – and this is from whence the drive feeding our reliance on Technology comes from.

¹⁷² This existential concept was discussed in Chapter Three (see section: *Sartrean Fear and Anguish (Angst)*).

¹⁷³ This existential concept was discussed in Chapter Three (see section: *Sartrean Fear and Anguish (Angst)*).

Braidotti redefined the monster as “a process without a stable object”, pointing out that “the monstrous other keeps emerging on the discursive scene, [and will persist] in haunting not only our imagination but also our scientific knowledge-claims” (1996, p.150). She explained that this is because

[d]ifference will just not go away. And because this embodiment of difference moves, flows, changes; because it propels discourses without ever settling into them; because it evades us in the very process of puzzling us, it will never be known what the next monster is going to look like; nor will it be possible to guess where it will come from. (1996, p.150)

The gene carrier – one whose body is anomalous – is the embodiment of difference and could be viewed as a genetic version of the monstrous body (monstrous DNA even!) within the teratology discourse.

We are pained by the knowledge that we cannot prevent the birth of the monster but, wherever possible, we seek to reduce our risk of doing so. We feel *Sartrean Fear* in the knowledge that we are largely at the whim of ‘Nature’/biology when we procreate: we may have difficulty conceiving; the foetus may have a genetic or chromosomal defect; the baby may suffer an injury during the birth, etc. Until recent history, human beings have been passive in relation to these possibilities. However, as a society, I contend that our reaction to this *Fear* has been to turn to Technology to provide solutions that will keep the threat of our lack of control over our genes at a distance from us. As individuals, then, we have an increased array of potential future reproductive decisions we can make – **our possibilities** – and whether we engage in them or not will be a matter for our own free will. In knowing this, however, we now experience *Anguish*. And this *Anguish* is exacerbated by the realisation that nothing other than our own free will actually prevents us enacting another possibility before us – to reject the Technology entirely and, by now doing this, actually embrace our *Despair*¹⁷⁴ rather than escape from it. Another way of achieving transcendence; for we remember “[t]here is no fate that cannot be surmounted by scorn”¹⁷⁵ (Camus, 2004b, p.490-491).

¹⁷⁴ This existential concept was discussed in Chapter Three (see section: *Despair*).

¹⁷⁵ This quote was also cited in Chapter Three (see section: *Absurdity and Revolt*).

CHAPTER SEVEN: EVADING GENETIC THREAT

INTRODUCTION

Because of our *caring*¹⁷⁶ engagement with the world, our genes and the genes of other human beings are an 'issue' for us. Our genes matter to us. The participants in this study confer on the carrier state its significance in light of their own self-defining Choices¹⁷⁷. It is due to them and those around them that it acquires any significance at all. We cannot presume to make a generalised judgement about what it means to people generally as it will always depend on what is important to each individual – their own self-defining Choice. How do they live-with the risks inherent in reproducing? How willing are they to accept these risks? What lengths will they go to in order to minimise these risks; to evade the threat? According to Armstrong *et al.* (1998, p.1657), the genetic counselling consultation has become a forum whereby counselees look to explore the possibilities for evading the web of genetic threat they find themselves enmeshed in. They wonder about avoidance through removing the threat: “[is] treatment, even cure, a possibility?” (Armstrong *et al.*, 1998, p.1657). The hope in Technology is initiated.

As human beings with the mindset that we are at the centre of a universe that is filled with beings which exist exclusively for our purposes, we no longer tolerate the idea that we or other 'thinking things'¹⁷⁸ should have to suffer. As our absolute Faith in religion has waned, so has our acceptance of the notion that our suffering is 'all a part of God's plan'. If there is no meaning to be found in our suffering (as is the case with all *absurd* suffering), we cannot abide the thought that we should have to endure it. And so, in our 'western-world-way', we show audacity in our dogged adherence to the belief that we can eliminate it. Perhaps because it is unsettling to be caught in the deterministic processes of nature and because we

¹⁷⁶ The existential concept of *Care* was discussed in Chapter Three (see section: *The Technological Attitude and Care*).

¹⁷⁷ The 'self-defining Choice' was discussed in Chapter Three (see section: *The Technological Attitude and Care*).

¹⁷⁸ Well, those just like us at any rate!

grapple with the fact that yet we still have total *Responsibility* for ourselves, the combination of our *Fear* and our *Anguish* drive us to Technology. We seek comfort in its embrace.

The lure of Technology is that it appears to maximise our possibilities, thereby enhancing our *Freedom*. However, simply being in a position to make a choice is not in itself a sign of *Freedom* as the individual is still constrained by the situation. Action must be taken within the given situation and there is no choice but to do this as **even lack of action is action**. In not opting for carrier testing and prenatal testing for CF, the 'at-risk' couple still makes an active choice. With the genetic and prenatal testing technologies that are available now, **there is no choice when making reproductive decisions but to choose**. Would-be parents cannot make reproductive decisions that are unencumbered by Technology; even their rejection of Technology amounts to a choice they were forced to make in relation to it.

This last analytical chapter examines how people look to Technology in order to evade genetic threat and considers the limitations of Technology in countering that threat.

Can the faith, hope and trust that families affected by genetic threat speak of in relation to the wonders of Technology be fulfilled? What do the dilemmas and paradoxes inherent in Technology's offerings look like in the genetic counselling/testing context? How do people perceive and manage genetic threat differently? Does Risk prevent parents getting the 'customer service experience' they would like from Technology? Are we working to ensure that genetic testing technologies serve to provide our children with more open futures, rather than more constricted ones?

TECHNOLOGY

The Paradoxes and Dilemmas of Technology

'BLIND FAITH' BRINGS (CERTAIN) HOPE FOR THE FUTURE

It is clear that the promise of Technology brings hope to many families who live with genetic threat. Their faith in the beneficence of its future offerings is reminiscent of the religious kind – certain, yet unable to be substantiated when the basis for it is challenged. An exploration of one mother's 'blind faith' and hope is summarised here.

Stacy revealed that she “definitely” plans to tell her daughter, Kristy, herself that she is a carrier when she gets older but, unprompted, she went on to minimise the impact she thought this would have on her daughter's life:

I just figure technology is so...advanced, that it's not really going to be such a big issue when she gets older – that's what I'm hoping anyway. (6: 29-30)

However, even if Technology could help her daughter minimise the chance of having a child with CF, her carrier state could still be a major issue for her if a future partner also turns out to be a carrier and they have to make a decision about prenatal testing. Stacy's 'hope' did not seem to encompass this so I asked some probing questions in order to discover what her faith in Technology is based upon:

Interviewer: *In what way do you mean that technology is advanced?*

Stacy: *That just... [struggles]*

Interviewer: *What sort of technology?*

Stacy: *Like testing the boy that she's going to be with, you know, the results will come through quicker and hopefully, you know, if she is a carrier and her boy's a carrier that there's...[thinks]...I don't know – better ways of treating cystic fibrosis – I don't know. I just hope for her that she doesn't have to go through any of that.*

Interviewer: *[...] [S]o you think that the therapies for CF will be a lot better?*

Stacy: *Oh definitely. (6: 33-46)*

The flow of conversation here is very interesting. Initially, her faith in Technology's advancements is the basis for her hope that her daughter's CF carrier status will not be a 'big issue'. From this we must presume she is confident that Technology will ensure she avoids the birth of a baby with CF. However, even if Technology means risk of the birth of a baby with CF can be largely eliminated, if she requires pre-implantation or prenatal diagnosis to do this it can hardly be said that it will be a 'small issue' for her to deal with. When I asked Stacy about the technology, her initial focus was on the carrier testing aspect of it but she paused before curiously changing tack to refer to it bringing "better ways of treating cystic fibrosis", thus admitting as a possibility the birth to her daughter of a baby with CF.

Surely then Kristy's carrier status has the potential to become a "big issue"? Is Stacy just avoiding the implication that Technology would allow her daughter to terminate a CF-affected pregnancy? And why would she avoid saying this to me given that she had already indicated she would have had a termination herself if her second pregnancy had been an affected one? To me, her faith in Technology is a vague one. She sees it as becoming so 'advanced', yet what this will mean for her daughter is not clearly established. I cannot help thinking that the better the therapies become for CF, the more ambiguous the severity of the disease becomes and the less clear-cut the decision for those who would consider terminating a CF-affected pregnancy on the basis of the child's anticipated quality of life. Surely this makes the decision harder. And while an optimistic outlook may continue to flourish as Technology progresses, with its gift to us being 'to test better and to treat better' it is paradoxical that this will only heighten the dilemma for potential parents.

Stacy's inability to clarify and pinpoint what aspect or particular advances in medical and/or genetic technologies she is relying on to curtail the impact her daughter's carrier status will have on her life is notable. Her belief in Technology seems akin to a religious one – based on a 'blind faith' rather than any concrete evidence; with a vague notion of what it will mean to her daughter, rather than a specific one. It is

striking that her **certain hope** for the future appears to be based on **uncertain premises**.

Because I was by now musing over the quandary that Technology lands people in, I asked Stacy if the optimistic outlook for CF sufferers (of which she spoke) makes the decision to terminate “less clear-cut”, “more difficult and complicated in some ways” (14: 21-26). She replied:

*[Y]ou’re probably thinking in the back of your mind, [...] you know, technology – I mean if I have this child now, you know, technology is so good that you know they’re going to be able to do something for my child and [...]... I don’t know. **There’s still a lot of people having kids with CF – they’re being born all the time.** I don’t know, but I just know that I wouldn’t have another one with CF. Definitely not. [emphasis added] (14: 32-37)*

The way Stacy articulates herself here is rather curious. She seems to imply that many people are choosing to proceed with known affected pregnancies (perhaps because of the optimistic outlook) but I doubt this is the case – I would guess that most CF births today are still totally unexpected, and to couples who have no knowledge that a CF gene mutation is in their family¹⁷⁹.

Stacy considers that the promise of continuing advances in treatment is one factor that may be involved in people rationalising their decision to continue with a CF-affected pregnancy but she is clear in her own mind that she would not have another CF baby. Viewed within the context of the rest of her interview, one would say this is because she knows that even if people with CF can one day live long and reasonably healthy lives, they are still likely to require a barrage of treatments and physiotherapy during childhood. She spoke of how emotionally and physically draining this is for parents and she had said she would not put herself through all of that again (13: 20-22). So despite her faith that Technology will bring a better future for those affected by the threat of CF, we see that she recognises this will not come soon enough for her.

¹⁷⁹ In fact, I have confirmed this to be the case – most babies with CF are born to parents who do not know they are carriers (J. Massie, Forbes, DuSart, Bankier, & Delatycki, 2007, p.721).

Stacy was not the only parent who expressed this kind of faith and hope in Technology. Where does it come from? Is it generally encouraged by health professionals who are at the interface of the wider public and the technologies? It has been said that while genetic counsellors are used to dealing with probabilities and uncertainty, physicians from other areas of medicine are not necessarily so comfortable dealing with uncertainty (Davis, 2001, p.13). Davis believes that they “tend to react by masking it with a false certainty” and she refers to, and then cites, another author Jay Katz (1984), who she says

talks about how doctors will acknowledge uncertainty when discussing scientific problems among their colleagues but mask it—even to themselves—when talking to patients. “They will acknowledge medicine’s uncertainty once its presence is forced into conscious awareness, yet at the same time will continue to conduct their practice as if uncertainty did not exist.” (Davis, 2001, p.13)

The machinations of Medicine and Science work to defend their respective technologies from the charge of fallibility, which serves the purpose of reinforcing our faith in it.

Novas and Rose (2000) suggest that the prospective consumers of these technologies are themselves building momentum behind the push for the actualisation of Technology’s promises. They say clinical medicine’s increasing constitution of the patient “as an ‘active’ subject – one who must play their part in the game of cure”¹⁸⁰ – has meant “[p]atients at genetic risk and their families [are no longer] passive elements in the practice of cure” (2000, p.489/490). Novas and Rose (2000, p.490) cite studies¹⁸¹ that they say show ill patients and others under genetic threat are “increasingly demanding control over the practices linked to their own health [...] and asking of medics that they act as the servants and not the masters of this process”. They explain that the patient is now consumer:

¹⁸⁰ Here, Novas and Rose cite the works of Armstrong (1984) and Arney and Bergen (1984).

¹⁸¹ They refer to the works of Rabinow (1999) and Rabeharisoa and Callon (1998).

These persons defined by genetic disease have an investment in scientists fulfilling their promises and discovering the basis of, and the cure or treatment for, genetic conditions. Medicine, including medical genetics, [...] has been one of the key sites for the fabrication of the contemporary self – free yet responsible, enterprising, prudent, encouraging the conduct of life in a calculative manner by acts of choice with an eye to the future and to increasing self well-being and that of the family (Novas & Rose, 2000, p.490).

Patients are changing: they are now making demands of Technology.

Duster (1990) has given attention to another source that feeds our hope in Technology – the media and its radically different coverage of the various ‘advances’ in medicine. As an example, he contrasted the treatment by the press of the results of two very different studies – a primary health care intervention in a lower socioeconomic area, which through information dissemination and prenatal education reduced infant mortality rates dramatically, versus the **possible** discovery of the chromosomal location of a genetic marker **possibly** linked to multifactorial clinical depression (1990, p.117). Duster need not have told us which of these made front-page news in the United States and our general experience of the media in Australia is no different – the exciting Technology-imbued advances always receive more attention. He also referred to a study by Nelkin (1987) that looked at the way the media covers science news and said it was noted that “the sensational big promises command attention, week after week after week” (Duster, 1990, p.117). Duster implies that it is “the medical establishment, premier research interests, powerful biotechnology lobbying, and insurance companies” that ensure this occurs, promoting their own interests ahead of expenditure on community health programs with proven cost-effective outcomes in “impoverished areas” where the “poor and uninsured” live (1990, p.119). A cure for CF and the prevention of the birth of children with the disease qualify as ‘sensational big promises’ by Technology.

'IN TECHNOLOGY, WE TRUST'

Trust in Technology can make it easier to face up to genetic threat in the here-and-now, and to live with the knowledge that one carries a disease gene and has passed it on to one's children. It can also mean people expect that gains made for CF sufferers to date will continue unabated; this improves their perception of the disease.

Kevin says that his and Anne's perception of cystic fibrosis has changed a lot during their affected nephew's lifetime. He attributes this to the positive outcomes that medical research has achieved for patients with CF:

I think [our perception of the disease has changed] because there's been an awful lot of research done on this. You know, the expected survival age has been going out and out and out, so when [our nephew] was born and when it was diagnosed – because it was a late diagnosis – like the early expectation was that he would probably die before he was twenty or so. Um you know, I think the expectation now is past forty sort of thing, which basically is a full life, well, relatively full life. By the time he gets there, it will be out further. Well, that's the expectation anyway. (8: 32-38)

His trust that Technology will continue to improve the prognosis for CF sufferers unabated is evident.

Trust in the promise of Technology also brought comfort to Kath after she found out that her grandson, Steven, had been identified as a CF carrier through the neonatal screening program. She recounted what her son and his partner told her being a carrier would mean for their child:

[I]f he married someone that's also a carrier then obviously their child's got a big chance of having cystic fibrosis. But they also said that by the time Steven's old enough to start having children, there will be a lot more known about the disease and it will be picked up a lot easier...so like the "shock horror" after knowing it wasn't quite so bad as not knowing what it was going to be. (3: 34-37)

So, for Kath, her trust in Technology brings an assuredness about the future, which lessens the “shock horror” of the here-and-now. She reveals she also thinks it will make it easier for her younger children to accept the news that they may be carriers, ending our discussion on this by saying:

And with the modern technology coming through with the babies being born now, there's a lot better chance of things picked up and that. Not that they can do a great deal about the disease – can they? – but at least they can be aware of it. (17: 54-56)

By Kath's own account, her confidence in the wonders of Technology even softened the blow of discovering that her son's CF mutation came from her side of the family rather than her husband's:

*I think at the time I didn't like that I'd done that but there's nothing that I can do to prevent it. [...] 'Cos I didn't know I was a carrier so... And at least now with technology increased so much, at least probably better now than having known say thirty years ago when I started having my kids – he's [her eldest son] thirty now. But there's a lot more that can be done and it probably would have shocked me more knowing that before I started my family. [...] **Just, at the time, there wasn't a lot known about it – or I didn't know a lot – and just to think that you could inflict the illness on a child, which is not a very nice illness – to knowingly give them. (10: 29-48)***

The irony here, of course, is that one cannot **knowingly** 'give' the disease to a child without the existence and availability of technologies that provide that ability to **know**.

Kath reiterates what she expressed in the passage above later in the interview, again crediting Technology with tempering the shock of finding out she carries a CF gene mutation:

I'm glad I know. I just think it was just “shock horror”, having six kids of my own and, like I said, to Dr [Surname], I said “Steven's been tested and he was a carrier” – so it had my GP a bit shocked and that shocked me too! [...] But um, finding out now probably is not quite such a shock as years ago because technology's improved so much. (21: 29-37)

Again, the paradox in her attributing a lessened shock to Technology's improvements is that it is purely because of these improvements that she has had to face this 'shock' in the first place. While knowledge of the laws of Mendelian¹⁸² genetics has meant there have been situations in which people became aware of their carrier status prior to the advent of genetic testing technologies, this could only be the case when an instance of the disease appeared in a child – and no one in Kath's family has the disease.

Kierkegaard did, after all, contend that the human condition is paradoxical!

TECHNOLOGICAL ADVANCES MAY BRING REGRETS

One mother highlighted the dilemma that the expectation of Technology's impending cure for cystic fibrosis brings for would-be parents.

Stacy speaks hypothetically about how her daughter may feel if she terminated a CF-affected pregnancy only to discover later on that had she had that child it would have grown up to lead a long and healthy life:

[I]f she does have a termination she might regret it later on. Because of the technol... What happens if she has a termination and, you know, like five years down the track they find a cure for cystic fibrosis? You know, that would probably really devastate her as well. I don't know. But I figured if you don't have to go through having a sick child, why should you, you know? (13: 47-51)

So while Technology brings options for parents, Stacy indicates that it has the potential to bring quite negative emotional consequences as well. However, she tempers this with: "But I figured if you don't have to go through having a sick child, why should you [...]?" "Why not choose a 'normal' life if Technology makes it available to you?", we almost hear her say.

¹⁸² It was Gregor Mendel who first formulated the idea that genes exist (though he did not use the word 'gene') in 1865. He inferred this "by observing certain progeny ratios in crosses between hereditary variants" (Griffiths, Miller, Suzuki, Lewontin, & Gelbart, 1993, p.19). His work constituted the prototype for genetic analysis as he was the first to propose that "characters are determined by discrete units that are inherited intact down through the generations" (Griffiths et al., 1993, p.20).

She talks about the improving prognosis for CF patients and speculates that even if Kristy does have a child with CF that the scenario would be much better for her as a mother than what Stacy had to face when her son was diagnosed:

Stacy: [W]hen Bradley was first born and we sat down and talked to the doctors, they said to me that, um...that fifty percent live – die before their early adolescence, and the other fifty live till their adolescence – I mean live till you know like eighteen or something. So I was devastated; I thought my child was going to be dead by the time it was ten, eleven. So I mean, they're not going to tell her that because things are different now.

Interviewer: So you think there's a more optimistic outlook?

Stacy: Yeah, and who knows in ten years time, you know what...it may be more optimistic, I don't know. I don't know, it just depends on technology – I've got no idea. (14: 10–19)

So again we see her optimism is dependent upon Technology – a faith that things will not be as bad for her daughter should 'the worst' happen.

Stacy's reference to Technology and what it may be able to do for CF patients highlights a matter that parents may contemplate when deciding whether they should terminate an affected pregnancy or not – the issue of how much medical technology (i.e., treatment and understanding of CF) may improve and how well their child could potentially be. The fact that CF patients are living longer and with a better quality of life, and that at some point in the future gene therapy may be successfully used to eradicate the symptoms of CF altogether, complicates the decision for would-be parents who would consider terminating a CF-affected pregnancy. The prognosis for CF patients today is ambiguous because of the array of treatments available and their increasing efficacy. One wonders how many parents may have continued with a CF-affected pregnancy because they feel a cure is 'just around the corner'. Certainly the hype surrounding medical and genetic research promises hope to parents and patients but how many believe a cure is closer than it is – and how much has the hype been used to promote this kind of thinking in the interests of securing additional research funding?

UNIMPRESSED BY THE PROMISE OF TECHNOLOGY

One CF 'survivor' was very proud of her late sister's relationship to Technology, which in turn seemed to give her a feeling of worth and importance, and yet she is still little impressed by its offerings.

Wendy describes her sister as having been a "guinea pig" for CF research in the 1970s (17: 8-9). She says that ultimately her parents must have given permission for this to occur but that her sister "wouldn't have had it any other way" and she is very proud of this (17: 16-22/41-42), even describing her sister as a "pioneer" to me the first time we spoke on the telephone. She speaks proudly of her sister's relationship to Technology and the place this gave her in the medical community – and also the status this afforded to Wendy as her sister. Wendy was frustrated to lose this aspect of her identity when she moved interstate:

*My sister had some of the best doctors in Australia, [...] professor of gastroenterology. I never had to...I never ever wanted for doctors. When I came to South Australia the first time, I had to actually like fight my way 'round in the world for doctors – like I expected as soon as I mentioned my name, I expected them to know who I am! But they don't have a bloody clue who I am over here. They haven't got a clue [sounds hurt by this and indignant]. **Over home, mate, I could just walk into a hospital and mention my name and it'd be like out come the red carpet...and all that sort of stuff.** (18: 45-52)*

Losing that feeling of importance within the clinical CF community was something she found herself needing to adjust to:

[N]o, it's alright not having doctors [know/acknowledge me]... I don't care really – you have to get used to it. (19: 37-38)

Despite this, it could hardly be said that Wendy is enamoured with Technology. Her devastating experience of losing a brother and a sister to cystic fibrosis means that she never had the option of having the same faith in Technology that Stacy has and she expresses little excitement about the 'promise' of Technology today. She remains relatively unimpressed with what the medical advances around CF

treatments have to offer sufferers; in contrast to Kevin (see section: In Technology, We Trust), her perception of the disease has not improved over time:

Nothing positive out there really [said quite definitively]. Not the way I look at it. Still nothing positive really. The only thing positive I heard of was an experiment – there was a woman in America that had a lung...had the whole transplant – and cystic fibrosis, the abnormality, didn't come back to her lungs. (16: 10-13)

Bereft of the faith and trust in Technology that other participants have, she does not share the same sense of optimism that things are improving for CF sufferers and that things will be better for the next generation of children should 'the worst' happen:

I do not want any – any – child of mine to have to suffer like I suffered as a child, and I don't want any of my grandchildren to ever, ever, ever have to even come close to suffering like I had to suffer as a child, especially not at the hands of a disease that I don't see in the next five to ten years them finding a cure for, other than transplants. And to me, that's not...unless they can show me something positive, then I don't want to know about it. (24: 36-41)

So thoroughly unimpressed is she by the current 'advancements' on offer in treating CF, we see that Wendy would not encourage her children to take the risk of going ahead with a CF-affected pregnancy only to place their hope in Technology.

Free to Choose but not Free not to Choose

In one sense choice is possible, but what is not possible is not to choose. I can always choose, but I must know that if I do not choose, that is still a choice. (Sartre, 1975, p.363)

ONUS TO 'USE' THE TECHNOLOGY

Some parents expressed the idea that not to 'use' – that is, not to act upon – genetic information is to ignore it. To them, it behoves prospective parents to fully take advantage of genetic testing technologies simply for the fact that they

exist and are available. Parents become blameworthy in their eyes if they opt against testing and then have a child with a 'preventable' genetic illness.

Kath reveals her attitude that people may as well face up to their carrier risk, take advantage of the technologies available and be carrier-tested if they have a CF mutation in the family:

I don't really know if my brothers and sisters should have got checked. I don't really know – that's up to them, I suppose, but if I was them I think I would rather be tested and know than just ignore it. But everyone's different, aren't they, how they react? (17: 20-24)

In line with this, Kath would like all of her children to be carrier-tested. Whatever the outcome, she sees that it will be beneficial – if they do not carry the familial CF mutation, it will be a relief to know that; if they are carriers, they will know it is advisable to have their partners tested (11: 29-31):

[W]ith today's technology, it's worth having the testing just for relief of mind – to know that either you haven't got to worry 'cos you're not a carrier or you are a carrier and it's better knowing about it than burying your head in the sand and not being interested (11: 32-36).

She says that testing will bring her peace of mind as a parent (11: 42-45). For Kath, it seems the very fact of the existence of this technology is *à priori* a reason to use it. However, in her eyes, it is also the way one should behave in order to be seen to be facing up to the genetic threat in the family.

In some of what Rose's Mum says we glean the attitude that the very existence of genetic testing technologies puts an onus on people to use it. She cites one case of a woman who has three children affected with CF who attend the same hospital as her granddaughter does for treatment:

When you see Susie or anybody else in her position in hospital and the things they go through, the drugs they have to have and all this sort of thing...you think, "Well, why put a child through that? Why knowingly, knowingly – 'cos I think there is one [mother] at the Children's Hospital that's got about three – three children with [CF].

She comes in and out. And as [my daughter-in-law] says, she just...she couldn't cope with more than one – even though, as I say, Susie is in reasonably good health.

[...]

[My daughter-in-law] apparently said to her...well, she said, "I wanted children". [My daughter-in-law] said, "But you knew with the first one". But she said, "I wanted children". Yeah, you want children – of course you want children but you don't put a child, knowingly, knowingly put a child through anything like that. (28: 32-44)

We see that Rose's Mum casts judgement against this mother's choice not to access the technologies that could have prevented the birth of her second and third affected children. Because of Technology, she is perceived as blameworthy rather than as unfortunate; she is seen to have **chosen** to bring these children into the world and is therefore seen as **responsible for their suffering**. This resonates with the reference to Clarke (1997c, p.85) made in Chapter One (see section: The Problem of Knowing: Population Screening For Genetic Carrier Status), that parents who have a child with a genetic disorder that severely impacts on their quality of life may be seen as irresponsible and blameworthy if it is known that they declined the technological options available to avoid their birth. This must be seen as an extra pressure for some parents to terminate rather than proceed with an affected pregnancy. So while more options may seem to bring greater choice for parents, this does not necessarily increase their *Freedom*.

The "ambiguity of choice", and its interplay with client autonomy in genetic counselling scenarios, has been discussed previously by Dena Davis:

[W]hile genetic testing is usually presented in terms of increased choice for women especially and prospective parents generally, in fact it may also represent a lessening of choice. Making choices is an emotionally exhausting and energy-consuming endeavor [...]. The more possibilities exist, the more it appears necessary to defend and argue for one's choice. (2001, p.16)

Given Rose's Mum's account, we imagine that the mother of three CF-affected children probably has had, and will continue to have, to "defend and argue" for her choice – much like Josie did when she chose, against her doctor's advice, not to have prenatal testing for CF (see section: Josie's Tale: "Oh, Here I Go Getting Another Blasting!"). Davis explained that while genetic testing capabilities may be "liberating" for some, or may indeed allow others "increased control over their lives [...] and more reproductive choices", there will be people for whom the technology is "oppressive" as it actually makes the decision to go ahead and have a baby with a genetic disorder a more difficult one (Davis, 2001, p.17).

The other point that Dena Davis would take up with Rose's Mum is her comment (quoted above) that it is wrong to "knowingly put a child through anything like that" – with 'that' being a genetic disorder that is likely to cause the child a great degree of suffering. Davis calls this ethical dilemma the 'harm conundrum' and says many writers deflect criticism away from parents who 'choose' to bring a child such as this into the world, arguing that

counter to our intuitions, no one has actually been harmed, as the child himself could not have existed otherwise than in his suboptimal state. Thus unless the child's current existence is so terrible that he would have been better off never having been born, he has not been harmed by being born in his damaged state. (2001, p.35)

Davis points us to a legal perspective provided by J. Robertson (1986, p.987) on the "protection of offspring" born via "noncoital reproduction". He says:

Risking damage to offspring would not seem to wrong the offspring if it were not possible for them to be conceived or born without undergoing the risk of damage. If the only possible means of conception or implantation also risks damaging offspring, no wrong to offspring occurs with use of that means. Avoiding the damage means avoiding the birth of the child. From the child's perspective, the risk-creating activity is welcome, since there is no alternative way for this child to be born. (1986, p.988)

This thought can also equally be applied to the child born with a potentially 'preventable' genetic illness to carrier parents today – these children can only exist if the parents engage in "risk-creating activity", that is, the natural method of conception followed by the rejection of reproductive technologies.

Rose's Mum says she wishes her son knew he was a carrier prior to the birth of his affected daughter because then he and his partner could have eliminated their risk of having a child with CF:

[T]hey could have done things differently, and Susie still could have been Susie but not Susie as Susie is today. [chuckles] She could have been an entirely different Susie! (17: 1-4)

It could be said that Rose's Mum takes up a position of *Bad Faith* here, denying that Susie can only ever be who she is – a little girl who was born with CF. In fact, Susie's reality is that she could either have been born with CF or not exist at all. The genes causing cystic fibrosis are an intrinsic part of her DNA – and had her parents taken steps to avoid the birth of a child with CF, they would have had a different child altogether, not Susie¹⁸³. The contingency of Susie's existence – that it relied on her parents not being aware that they each carried a CF gene mutation – is brought to the foreground here but not acknowledged by her grandmother.

There is yet another example in this interview of the expectation that people should act upon and fully utilise the genetic information they are in possession of. Rose claims that it is her son's choice what he 'does' with the information that he is a CF carrier:

[A]s long as I know that I've done everything I can...and I've told him and told him what to expect...but I think that he'd be smart enough to do something about it, not put himself through it or his children, with a chance of getting it (29: 51-53).

¹⁸³ The wording here is paraphrased from Dena Davis' ethical discussion of the couple who knowingly conceives and gives birth to a baby with a serious genetic disorder, rebutting the claim that this is wrong for the sake of the child itself (except in the most "horrible instances") because "it relies on the assertion that the child's life will be so terrible that it would have been better off if it had not been born" (Davis, 2001, p.37).

She says that she has brought him up talking to him about his cousin's CF:

[W]hen the time comes, I can say, "Now you've got all this information, you know what to expect – it's up to you now to go and hopefully do the right thing and be tested. Not just go willy-nilly and have a child. Have your wife tested and you can go from there with whatever result she comes back with." Rather than just ignore everything. (30: 4-8)

We see here that Rose has a very certain idea of what the "right thing" for her son to do is. We get the feeling that she sees herself as having done the right thing; she has been responsible and it is now up to her son to be responsible and avail himself of the testing technologies. This, combined with the level of investment we saw earlier in her battle to have him carrier-tested as a child, does not augur well for her son's ability to grow and make reproductive decisions that differ from, perhaps even conflict with, his mother's expectations.

This closed notion of a **logical** and **right way** to 'use' the information and technology that can prevent the birth of a child with a hereditary condition is not new. Davis refers us to the work of Sheldon Reed who coined the term 'genetic counselling' in 1947, defining it as the "unbiased presentation of information without guidance" (cited in Davis, 2001, p.14)¹⁸⁴. She said he actually

felt that there was little need for guidance, as parents would act "logically" and not have children if they had a high risk of genetic disease (Davis, 2001, p.14).

We have also been told that

[f]rom its very beginnings, many eugenicists, including the founder of the eugenics movement, Francis Galton, were opposed to coercion, believing that if people were properly informed they would naturally make the "right" reproductive decisions (King, 1999, p.177).

¹⁸⁴ In providing this information, and in directly quoting Reed, Davis references the following source: "Reconsidering 'Nondirectiveness' in Genetic Counseling", *Gene Letter* 1, 4 (1997); <http://www.genesage.com/professionals/resources/answercenter/geneticcounseling.html>. This information could not be verified as it seemed this webpage was "out of date" when I tried to access it in September 2008.

From many angles then it would seem that expectant parents are under pressure to have prenatal testing, not least because in the event they do have a child with a disability or illness they may anticipate being blamed by others for failing to take a test.

The results of a study conducted in three western European countries add weight to this (Marteau & Drake, 1995). The study looked at whether attributions for the birth of children with disabilities are influenced by the widespread availability of prenatal testing for foetal abnormalities; its findings suggesting that “both health professionals and lay groups make judgements about women’s roles in the birth of children with disabilities” (p.1130). The results showed that

screening history of the mother was the single most important factor influencing attributions of control and blame following the birth of a child with Down syndrome. A mother who declined the offer of testing was seen as having control over this outcome, and was in part blamed for it. (p.1127)

In relation to reproduction, people are becoming less and less able to claim a standpoint of reluctant *Despair* – that is, that the outcome is ‘out of their hands’. Advances in genetic testing technologies mean prospective parents are increasingly seen to be in control of what they once were not – the transmission of their genes.

SANCTIONING THE CHOICE OF INACTION

In opposition to those (grand)parents who expressed the idea that the onus will be on their carrier (grand)children to avail themselves of the genetic and prenatal tests that Technology can deliver, not all thought that knowing one is a CF carrier necessarily implies specific action.

On the topic of how she felt about finding out her newborn son carries a CF mutation, Vicki said:

*[Y]ou just keep thinking ahead – what about when Zane wants to have children and how do we tell him about it and **should it really impact on his choices anyway?** So I guess that was my main concern. [emphasis added] (13: 13-16)*

In saying this, she reveals that there is no assumption on her part that her son will take any one particular course as a result of knowing he carries a CF gene mutation. In fact, she questions whether it should affect any reproductive decisions he makes at all.

Likewise, Tanya does not necessarily assume that her daughter, Georgia, will have her future partner carrier-tested:

[I]f she decides that that's the person that she wants to be with, who am I to say, "Well, you should have him tested and if he is then 'no'..."? That won't work! [laughs at the ridiculousness of that idea] (19: 28-31)

Probably because of her own anti-abortion stance, she sees the only 'use' her daughter may have for carrier testing as being to determine whether she wishes to stay in a relationship with a man who is also a carrier of a CF mutation. However, it seems the notion that she would actually encourage her daughter to take this path strikes her as fairly preposterous. She says it little matters whether Georgia's future partner is carrier-tested or not if, as a couple, they decide they definitely want to have children and would not consider terminating a pregnancy:

I see that it doesn't really matter. I think, you know, that it may be useful for them to know that but then again it may not be an issue to them. I think it kind of would ride on what they think about it and not what I think about it. (19: 39-41)

All in all, she seems quite nonplussed by the availability of genetic and prenatal testing technologies. This bodes well for Georgia's right to an open future, as the maximum array of possibilities remain intact before her.

Confronting Risk

HEIGHTENED AWARENESS OF RISK

Two couples spoke of the way that medical/genetic surveillance during the prenatal and neonatal period brought risk to the foreground. Once there, it remained – and this heightened awareness of risk saw them more acutely conscious of all the potential threats to their children’s health in the years to come.

During her pregnancy, tests had indicated that Leanne’s baby girl may have been affected with Down’s syndrome (3: 16-17). She felt that her “cautious”, non-“over-excited” approach (15: 40/44) to coping with this risk had served her well:

I didn’t get really, really upset about the Down’s syndrome thing [...] just played it really calm [...] and we went and had the ‘amnio’¹⁸⁵ and everything was fine. (3: 23-25)

Even though the tests were negative for Down’s syndrome, this early-on encounter with risk meant Leanne “protected [herself]” throughout the pregnancy and prepared herself for the chance “something still could be wrong” (3: 18-20); she “never really let [herself] get excited [about it]” (6: 36). Her husband, Rob, said too that it was not until “tests started happening and results started to come back” (7: 6) that he ever considered “those sorts of sides” to pregnancy (7: 5) – that “you don’t just get pregnant and have a baby ‘cos of all these things to do along the way” (6: 46-47). Through his encounter with the ‘technological gaze’ (a concept discussed by A. Robertson, 2001, p.301), Rob now sees pregnancy as a **process** whereby one anticipates Risk and actively seeks to evade its threat. We note that the “‘at-risk’ consciousness”¹⁸⁶ (A. Robertson, 2001, p.300) has emerged and Rob

¹⁸⁵ Colloquial term for ‘amniocentesis’.

¹⁸⁶ The notion of risk is now central to discourses related to individual health in western society and it is from within these discourses that the ‘at-risk’ consciousness has emerged (A. Robertson, 2001). According to A. Robertson, “such consciousness contributes to the emergence of a particular form of subjectivity – that is, a particular way of thinking about, relating to and situating the self in terms of the broader social and political context within which the self is embedded/located” (2001, p.300).

has adopted the “calculative and prudent attitude with respect to risk and danger” that characterises the “individual-as-enterprise”¹⁸⁷ (Petersen, 1996, p.51).

Leanne went on to explain why, having protected herself, it was then so difficult to deal with the unprepared-for news that her daughter had a positive result in the Guthrie test and may have CF:

*I think what really hit me with this was once we had it, it was like everything's going to be okay now. Like I've got through the pregnancy. I've got her. She's fine. There isn't anything wrong and I think that's why this really affected me more than the first testing with the Down's syndrome. It's because I suddenly thought, "Oh God, like I thought everything was going to be okay now". (3: 26-31)
[...] [T]hat's what I found the hardest – that I had been so guarded and then once I'd had her, everything was...I felt, you know, you get that false sense of security. (3: 49-51)*

As long as Leanne had remained conscientious about managing the risks confronting her, she feels she was better placed to cope should anything have happened. It was in the moment of letting down her guard – **forgetting Risk** – that she was less emotionally equipped to handle another threat to her daughter's health and happiness. In accordance with the adage 'once bitten, twice shy', Leanne's consciousness of all that can go wrong in the life of a child has been intensified by her own sharp encounter with a once-very-real threat to her baby:

And now I think, after having that test done and the possibility that she could have it, has made me just more aware that any time there could be anything down the track that, you know, you never know when she is going to be diagnosed with cancer or leukæmia or, you know, anything. So I think, for me, that's what really sort of like...“Oh, we're not, you know, out of the woods – it's always going to be forever something that it could be”. (3: 49-56)

¹⁸⁷ Novas and Rose have provided an insight into the 'entrepreneurial subject', for whom "life has become a strategic enterprise" (2000, p.487). They say that Medicine has been "one of the key sites for the fabrication of [this] contemporary self – free yet responsible, enterprising, prudent, encouraging the conduct of life in a calculative manner by acts of choice with an eye to the future and to increasing self well-being and that of the family" (2000, p.490).

The experience she describes here suggests a kind of 'lost innocence'. Medical surveillance, rather than illness, has heightened her awareness of risk and seen her become more attuned to her (*Sartrean*) Fear.

Anne and Kevin see that it was a fairly 'close call' they did not have a child with CF, presumably because Kevin had a one-in-two chance of carrying the gene; they did not opt for carrier testing prior to their first and second pregnancies; and, when they did, it turned out that Anne was a carrier (although Kevin did not turn out to be):

We feel like we sailed very, very close to the wind [by not having a child with CF] but in the end [...] it wasn't, "Oh, thank God that's it" because you're just so much more aware of how dangerous it is. Some parts of life are very, very dangerous and, "Don't be too relieved about that, guys, because there's other things that come around the corner, obviously unannounced and 'clonk' you!" [speaking very quickly]. I guess it just...maybe it reinforced a fatalistic approach at life. (Anne, 26: 56 – 27: 5)

Like Leanne, Anne too remarks that a close encounter with genetic threat has heightened her awareness of all of life's risks more generally. Kevin supposes at the reason we are so startled when personally confronted with genetic threat:

Genetic diseases always strike other people. I think that's sort of an in-built sort of psychology in the human psyche or something. (26: 29-30) [...] Maybe it's part of the human survival trait, I don't know. (26: 40-41)

Interestingly, he looks to a genetic explanation for how we feel in relation to our genes!

TIM'S PERSPECTIVE: "IT'S JUST SUCH A SMALL CHALLENGE AHEAD OF OUR CHILD"

In contrast to the parents who felt that their close encounter with the CF risk had brought other potential risks to their child's health and well-being to the foreground, one father used those risks to put this particular genetic threat to his son into perspective and to quell his concerns about CF.

Tim is a self-described “optimist” (23: 20) who manages to put the potential threat posed by one’s genes into perspective. He plays down any issues there may be in telling his son he carries the CF gene mutation by saying, “at the end of the day, what he does have anyway, it’s not that uncommon anyway from what we were...what we were told” (20: 25-26). Both he and Vicki reflect on the fact that threat to the newborn can come from many angles:

Tim: There’s so many other diseases as well. Like, this is just one. Like, when you’re having a baby, really it’s only one of the many, many diseases that, you know, or genetic problems that we could have had anyway.

Vicki: Or even things that can go wrong...

Tim: Or things that can go wrong just through childbirth.

Vicki: Who we’re having contact with, you know, viruses and toxoplasmosis and all sorts of things. (20: 51 – 21: 4)

Tim confided that they did not have “a real piece-of-cake pregnancy”, having had a few ‘hurdles’ to get over in both the ante- and the postnatal periods (22: 31-35). In a similar way to Leanne (see section: Heightened Awareness of Risk), he described them feeling that the CF threat came along just when they thought they were ‘out of the woods’ and for this reason it “seemed a bit surreal” (22: 35-36), however, he still managed to see a bright side to their situation:

I think at the time, cystic fibrosis was something that was very scary but it was nowhere near as scary as some of the other...the other problems that some babies are born with. You know, we’ve got friends whose babies were born with a hole in their heart and things like that, so... (22: 54 – 23: 2)

In response to Vicki reminding him here that CF is a terminal illness (23: 5), Tim alluded to his childhood friend who he has seen grow up with the disease, live into her late twenties against all expectations (21: 34-42), and still have what “seems like a really good quality of life” (24: 1). Due to this experience, he said it has “never really sunk in” with him that it is terminal (23: 21-22) and he feels he tends to “[look] on the positive side” when it comes to CF (23: 53-54).

Tim confirmed that knowing only Vicki is a carrier (and not him) has reassured him in relation to their own future pregnancies “because the risks with pregnancy are so high anyway” and it is just another one of those risks (25: 19-21). Speaking about his son’s genetic situation – that he has a chance of having a baby with CF one day – Tim says:

It’s hard to picture like what will happen in, you know, in the current state of how the world has progressed just in my lifetime (which is 30 years). It’s hard to picture where the world will be in ten years time and what, you know, what will face him. CF, you know, the way the world goes nowadays, CF’s probably gonna be the least of his concerns, just with the other diseases and what’s happening in general life. It’s hard to picture. It’s just so...it seems so far away. (25: 44-49)

He sees the fact his son’s future partner may also turn out to carry the CF mutation as “such a small challenge ahead of Zane” and, to illustrate his point, compares it with the risks that were posed to him and his four brothers growing up as children on a farm:

*I grew up like in a family of all boys and we grew up on a farm and like if you look at generally, you know, the risks associated with kids, I suppose, are fairly high on farms and I look at all the things that have happened in the time that I’ve been alive, you know, with my brothers being allergic to bees, like the number of times that they’ve been critical and just in accidents that we’ve had on the farm – motorbike accidents and car accidents that I’ve been in – you just think that the one-in-25 or whatever, the one-in-100,000 (sic) chance that you might contract CF seems like **it’s just such a small challenge ahead of Zane**. There’s so many other things that as parents you are going to have to protect him from, probably before that, which is probably more scarier than anything else. (28: 23-33)*

His words remind us that an over-focus on genetic information and Technology can skew our perspective – after all, we have many other things in life to worry about!

A. Robertson has argued that “current discourses on risk both make possible and are made possible by particular diagnostic/screening technologies” (2001, p.293). She suggests that

[w]ith the current push to make genes the ultimate ‘risk factors’, the individualization of health, and health risks, is complete: health risks are located in the very ‘building blocks’ that make up the person, not in the social, physical or political environment. (2001, p.301)

We see here that Tim does not take up the prevailing biomedical discourse on genetic risk, showing there is potential for the geneticisation of risk (Lippman, 1991) and the “‘technologization’ of risk” (A. Robertson, 2001, p.301) to be resisted.

The Limitations of Technology

For the existentialists it is neither external political circumstances, nor a lack of technological knowledge, nor a want of wisdom, nor an imperfect moral development of the race which prevents the attainment of human happiness. It is the human condition itself which does so. Man could not become happy without ceasing to be man. (Olson, 1961, p.15)

TECHNOLOGY DOES NOT ELIMINATE RISK

One mother expressed great annoyance that carrier testing does not definitively rule out the presence of a CF gene mutation – she expected more from Technology! Some spoke of the inevitability of risk in the reproductive arena, but also the specific risks that Technology itself brings to it. Each assessed the risk that prenatal testing technologies posed to their unborn child in their own way – some were prepared to accept it and others were not, depending on what threat they were most concerned to evade at the time.

Wendy was “rapt” after a DNA test showed her most recent partner did not carry any of the CF gene mutations routinely tested for (22: 21) but she expressed frustration that they could not be told definitively that he is not a carrier:

[A]s they say, they can't rule him out...being a carrier [...] [J]ust done six week's worth of bloody genetic testing [sounds frustrated] and you can't say that...even though it says 'no', they say that it still doesn't eliminate them and I think "Ohhh" [frustrated because she perceives the technology as inadequate]. [...] I mean they've done six week's worth of genetic testing and either you're a carrier of the bloody disease or you're not – simple, whether it's a common mutation or not. It's either you are or you aren't – the way I see it. (16: 38-50)

Her incredulity about this reveals that she does not adequately understand the way the testing works – there are hundreds of variations of the CF disease allele and it is only practical that the few more common ones be tested for. The fact he has been shown not to carry any of these tested-for mutations means he has a highly reduced risk of being a carrier but that it cannot be ruled out completely. Wendy is correct in saying:

[I]t still never eliminates the fact that I'm a carrier and there's still that one chance in 400 that I could still produce that one baby! So...I don't know...I don't know how anyone else feels about that. But I don't know...that was still probably in the back of my mind. (22: 21-24)

The risk of conceiving a baby with CF lingers and, in this circumstance, prenatal testing would not be efficacious because any less common CF mutation would still not be detectable. Wendy's expectations of Technology are high: eliminate all risk; leave nothing to chance. She refuses to accept the fallibility and frailties of science; the inadequacies of Technology ... yet she has to live with it.

In stark contrast, Anne and Kevin claim they have always accepted the risks inherent in procreating. They knew that the CF gene mutation was in Kevin's family four or five years before getting pregnant but they “didn't think about that” by the time they wanted to start their own family (4: 22-23). They did not look to Technology to

evade this particular threat. On this and other risks attendant to having children, Anne said:

I guess we hadn't sat down and worried about all of those. Again, even going back to before [our first daughter], we hadn't sat down and done it as a scientific, you know, these are the risks for having children, let's worry about them beforehand. [...] I mean we knew they were there, we'd done enough reading and we were well-educated enough to know that, again, that it's all possible, but we didn't individually worry about... I mean, it's the same thing – what do you want as a baby? Just a happy and healthy child. It's just that you have to have an optimistic view about what's going to happen and if something happens then you deal with it afterwards. (17: 13-24) [...] [L]ife is full of risks and you just... I guess there's a point at which if you decide to have children, you have to be accepting of some of those risks. And I think if you didn't – if you weren't able to accept that – then the whole pregnancy would be miserable. (17: 35-40)

We see that Anne and Kevin were able to accept the 'givens' of having children (facticity) and thus lived comfortably with *Sartrean Fear*. However, this changed for Anne after their second baby, who had Down's syndrome, was stillborn. Prior to this, she had never "[felt] the need" to go through the whole gamut of prenatal testing (20: 52-54) but the thought of enduring a pregnancy consumed by "the fear of 'What if? What if? What if? What if? [...]'" (20: 34) changed this:

[My first] pregnancy, I remember with huge joy. And of all the things I was worried about, once [our second baby] had died, is that I'd never have another joyful pregnancy and it was very hard (because I was still grieving that year) but I was determined to try and understand what was happening and reduce as much risk as possible – well, be informed about whatever risk I was facing and then come to the point where I could try to enjoy and look forward to her birth, I guess. [begins to sob] (17: 44-50)

Despite never having arrived at a decision as to what she would do if tests did come back positive – she "couldn't make a theoretical decision based on that sort of information" (21: 17) – Anne was determined to have the testing this time around. Such was her need for certainty and her feeling that "the earlier I know, the better

‘I’ll be’ (21: 14), she was prepared to undergo chorionic villus sampling – “almost twice as risky” as amniocentesis (21: 13) – so she could have her results earlier¹⁸⁸.

We see that it was less so genetic threat (e.g., Down’s syndrome and CF) that Anne was seeking to evade by turning to Technology than emotional threat. After her devastating experience, it was the **fear of anticipating genetic threat** for months, **rather more than the genetic threat itself** which terrified her. But Technology is fallible. It is imperfect. It does not eliminate all risk and, in fact, it often actually introduces risk. Some people accept these risks and some do not. While Anne accepted the risk posed by testing in her desire ‘to know’, Rose and Josie were unwilling to do so.

Rose considered undergoing prenatal testing during her first pregnancy but decided against it because of the 1% miscarriage risk associated with amniocentesis:

It was awful that you had to wait the full nine months to find out whether [he] would have had it or not...yeah, it was awful. I mean, yeah I could have went and had the ‘amnio’ but the chance of losing him was even worse, I thought, for me. (5: 40-42)

Unable to utilise the available testing technologies because the risk of doing so was unacceptably high to her, she described feeling “anxious and nervous” (5: 56) throughout the pregnancy. Without knowing whether the baby’s father was a carrier or not, she said it felt more like “a 50:50 chance”¹⁸⁹ that her baby would be born with CF (6: 1-7). She explained that a large part of this was because the father was from England and she remembered being told that “it’s from English and German descent” (6: 11-12). She said, having read ‘leaflets’ during her pregnancy that indicated there is a one-in-four chance of the birth of a baby with CF where

¹⁸⁸ According to the Joint Human Genetics Society of Australasia/Royal Australian and New Zealand College of Obstetricians and Gynaecologists Prenatal Diagnosis and Screening Committee, due to the risk of adverse outcomes, it is not recommended that chorionic villus sampling be performed before ten weeks gestation or amniocentesis routinely performed before fourteen weeks gestation (“Prenatal Diagnosis Position Statement,” 2006).

¹⁸⁹ In fact, without knowing the father’s carrier status, the actual chance the baby would have CF was approximately one-in-100.

both parents are carriers, “automatically I just thought ‘oh, 50:50’, that’s it. [...] [T]hat’s just how I thought” (6: 21-25).

Once pregnant for the second time, Josie also weighed the potential benefit of prenatal testing for CF against the risk. Particularly because she had decided against terminating if the foetus was affected, she could not see that it was worth increasing her risk of losing the baby just to know whether the baby had CF in advance of the birth. While a baby with CF benefits from early treatment, it would only be a matter of a two-week wait to find out because hospital staff would organise testing of the cord blood as a matter of priority. She also considered it untenable in her own particular circumstances to minimise the risk as much as others may be able to:

I weighed it up and I thought, “Well, is it...(oh, so much percentage of miscarrying)?”. And I thought, “Well, a high percentage is gonna be quite higher because I’ve got two-year-old twins!”. I thought, “I’ve gotta have bed-rest for 24 hours”, like that was one black mark against it (or, how long it said bed-rest). [...] I thought, “Yep, right, how am I going to do that with a CF child and two-year-old twins?!”. [we laugh at the implausibility of the idea] And I thought, “Well, there I am already putting myself at risk for having a miscarriage because I’m not going to be able to [rest]” – I can say that right now! (18: 22-32)

Given her aversion to abortion, a lack of available support, and her wish to protect her unborn child from potential harm, we see that Technology had nothing to offer Josie here – much as it was unable to protect Rose from nervousness as she anxiously awaited the birth of her child.

TECHNOLOGY DOES NOT ELIMINATE SUFFERING

One mother highlighted eloquently one of the paradoxes of Technology: in its concern to eliminate suffering it invites another kind of suffering.

Not once in her interview did Cindy express hope that Technology would help those with CF, now or in the future – nor would she “be presenting an optimistic point of

view” to her children about what it would be like to have a child with CF (10: 49-50) despite the treatments that have been developed since her brother died. Between the years 1973 (the death of her brother) and 1984 (the birth of her first child), Cindy said she did not “reckon a lot happened” in terms of genetic research – the relatives of children with CF were still unable to find out for sure whether they carried the CF gene mutation or not (13: 53). This meant she had only one choice to make in terms of managing her ‘CF risk’ – to accept the risk involved and go ahead and have children or to decide against trying to have children at all:

I guess I was positioned as still thinking, “Well, you either do or you don’t. You run the risk. You don’t...you can’t find out if you’re a carrier. All you can do is find out if you have a child who has CF”. So there wasn’t any room there to go anywhere beyond that. So unless you just didn’t want to have children at all because there may be a risk, [pause] nothing you could do. (14: 15-19)

And indeed, she prefers that this was her choice as opposed to having been in the situation of needing to make a decision post-conception:

***Cindy:** I was glad I wasn’t faced with knowing that I had a...that I was carrying a child who had CF. That would have just been devastating. So...haven’t been there.*

***Interviewer:** Knowing that you might decide to terminate or...*

***Cindy:** Or might decide to go ahead and, yeah... [have the baby]*

***Interviewer:** So really whether the technology is available or not, it’s still gonna be painful [a few words inaudible]...in a position really they are still affected by CF...*

***Cindy:** Oh yeah.*

***Interviewer:** Whether they have a child with CF or not.*

***Cindy:** Yep. Because **Technology is one thing and Life’s another**¹⁹⁰. (16: 30-44)*

¹⁹⁰ What a wonderfully insightful comment provided here by one of the informants in the study. So wonderful, it seemed the perfect note to end the presentation of data in this thesis on!

Is there a general belief that genetic testing technologies will help us to eliminate suffering? As with energy, which is neither created nor destroyed but only transformed, so too with human suffering. Technology's paradox continues: in our concern to put an end to one form of suffering, we create yet another kind. The difference between the two is that we make an active choice to bring 'technological suffering' into our lives but we seek to evade the '*absurd* suffering' that threatens our lives.

Camus once said:

Living with one's passions amounts to living with one's sufferings, which are the counterpoise, the corrective, the balance and the price. When a man has learned—and not on paper—how to remain alone with his suffering, how to overcome his longing to flee (the illusions that others may share), then he has little left to learn (as cited in Charlesworth, 1975, p.5)¹⁹¹.

And yet, these 'sufferings' which are the counterbalance to our 'passions' are the target without remittance of medical and genetic technologies today. In the drive for ever-increasing genetic testing technologies, we show neither recognition nor acceptance of our *absurd* fate and, for Camus, these are key to our ability to transcend the *Absurdity* of human existence.

The Myth of Sisyphus tells us that, having admitted to the cold indifference of the universe, it then becomes possible to extract an authentic sense of meaning from within it (Camus, 2004b). Succinctly summarised by Flynn, we are counselled that

our only hope is to acknowledge that there is no ultimate hope.
[...] [W]e must limit our expectations in view of our mortality.
(2006, p.48)

In other words, we must expect to encounter some sufferings in our lives. Camus says that "[s]eeking what is true is not seeking what is desirable" and that the

¹⁹¹ Charlesworth cites Brée (1972) as his source for this quote. Apparently, this passage of the text was found underlined in Robert Kennedy's own personal copy of Camus' *Notebooks* and it was in this context that Charlesworth cited it in his book.

absurd mind, rather than resign itself to the falsehoods and “roses of illusion” of religion would rather “adopt fearlessly” the position of *Despair* (2004b, p.470) – that is, facing up to the realisation that there is much that happens in the world about us over which we have no control. With the *Technological Attitude*, it is difficult for people to accept this and it brings us to increasingly look to control, or to have controlled, every aspect of our existence and the community and world around us.

In his book, *Backdoor to Eugenics*, Troy Duster (1990) problematises the justification of genetic testing technologies by their potential to alleviate suffering caused by disease. He claims

[t]he promise and achievements in recombinant DNA research at the molecular level have made us vulnerable to [...] a diversion of attention and resources from other solutions, although there is already evidence that we could make a larger and faster difference (and cheaper) in the reduction of human suffering (1990, p.126-127).

His fear that it will be “only a matter of time before elliptical eugenic uses are made of these new technologies” (1990, p.127), particularly with the complete mapping of the human genome, reveals a scepticism that human suffering is the only motive behind these technologies. While “the major banner for the Human Genome Project would be the prospective gains in health and medicine”, he expressed concern that commercial interests will most likely prevent some companies sharing all of the key information freely and called it “another costly exploration into potential and yet unknown health benefits while we already know enough to save millions by applying existing knowledge” (1990, p.127). In other words, the expansion of genetic testing technologies will come at the expense of known health solutions.

Lippman holds a similar view:

For society, genetic approaches to health problems are fundamentally expensive, individualized and private. Giving them priority diminishes incentives to challenge the existing system that

creates illness no less than do genes. With prenatal screening and testing in particular, the genetic approach seems to provide a “quick fix” to what is posed as a biological problem, directing attention away from society’s construction of a biological reality as a problem and leaving the “conditions that create social disadvantage or handicap ... largely unchallenged.”¹⁹² (1991, p.47)

It has been said that Lippman, Duster, and others argue that “genetic narratives of health and disease orient the ways in which problems are defined, viewed and managed within society”, and that this serves to “[redirect] scarce resources away from social solutions to social problems” (Novas & Rose, 2000, p.489).

CONCLUDING REMARKS

In the *Technological Attitude*, we see the world about us – the world of non-thinking things – as “a foreign territory, to be conquered and exploited” (Cooper, 1990, p.13). It is to the *Technological Attitude* that Heidegger, Buber and Marcel attributed our modern-day tendencies towards Technology and consumerism (Cooper, 1990, p.13). And it would seem that we have now turned this stance on ourselves. Our bodies, including our genetic make-up, have now also become territories to be conquered (and potentially exploited). They are now the subject of the ‘molecular gaze’, which is recasting life “as a series of processes that can be accounted for and potentially re-engineered at the molecular level” (Novas & Rose, 2000, p.487).

Geneticisation and genetic/prenatal testing technologies have transformed parents into consumers and their children into “consumer objects subject to quality control” (Lippman, 1991, p.23). This is heightened further by pre-implantation diagnostic techniques which have the potential to open up “an unrestricted free-market eugenics” in which children may be carefully selected by their parents “using the all-seeing eye of genetic technology” (King, 1999, p.180). King speculates that in a market such as this, children will no longer be seen as “a gift from God, or the

¹⁹² In this last sentence, Lippman cites McDonough (1990).

random forces of nature, but selected products, expressing, in part, their parents' aspirations, desires and whims" (1999, p.180). Their existence will become contingent less so upon the arbitrariness of biology and more so upon their parents' stipulated design. What then of *Nausea*?

This "manipulation of 'nature'" is one of the principal concerns troubling those involved in the "increasingly vigorous and public debate about the ethics of genetics" (Diprose, 2005, p.244). Diprose describes the "potential for designer bodies or gene therapy [as] a common cause for concern" (2005, p.244). She says that genetics is one of "the technologies of power deployed with the emergence of the modern biomedical and social sciences in the nineteenth century" that Michel Foucault collectively refers to as 'biopower' (2005, p.253). Diprose provides an insight into how these technologies work together to enact this power:

Without reference to law, and without displaying themselves as power, these sciences divide and assemble the body; evaluate, sort, and compare it. They thereby transform life by effecting distributions around a norm of health and well-being¹⁹³. The assumption of, and desire for, sameness pervades these sciences of the body. (Diprose, 2005, p.253)

Biopower, she says, is kindled by "this urge to recreate the self" (2005, p.253).

Biopower can be construed as a modern-day triumph of Technology. According to Flynn, Heidegger argued that "the triumph of the technical in contemporary society and the reduction of both nature and humans to mere 'resources' [are] the logical outcome of our forgetfulness of Being over the centuries and our desire to control", and it was this that culminated in the Nietzschean doctrine of the will-to-power¹⁹⁴ (Flynn, 2006, p.55-56). The will-to-power, as articulated by Nietzsche, was a direct and inevitable result of the death of God¹⁹⁵. Once He was proclaimed dead, human beings sought to replace one patriarchal system based on blind faith, belief,

¹⁹³ Here, Diprose cites Foucault (1978).

¹⁹⁴ This concept was discussed in Chapter Three (see section: *Freedom and Choice*).

¹⁹⁵ The significance of Nietzsche's announcement that God is dead was explored in Chapter Two (see section: *Reconciling the Christian and the Atheist Existentialists*).

obedience, submission, and hope for a glorious future without woe, with another: Religion was traded for Technology. With no external Keeper watching over our lives, determining our fates, we are left alone – *freed and forlorn*¹⁹⁶ – to take matters into our own hands. However, this also has meant having to take *Responsibility* for the outcomes of our choices; living with the decisions that we make; owning them.

The will-to-power, expressed at an individual and a societal level, leads us to do what we can to overcome our sense of powerlessness in the procreative sphere; a feeling of powerlessness that neatly sits within the concept of *Sartrean Fear*. Our determined attempt to gain control over the reproductive arena via Technology derives from an enculturated belief that some power over our individual destinies is available to us; a belief that manifests itself in our lives as *Sartrean Anguish*. This *Anguish* is exacerbated when we become the custodians of genetic information because we are coached, at the same time, to believe this will give us (and potentially our genetic relatives) control over our situation. Others too will now see that there is a responsible way for us to use this information. In this way, genetic testing technologies can be seen as an end- or evolving-product of man's¹⁹⁷ will-to-power – an expression of the desire for more effective control of procreation, parenting and populations.

¹⁹⁶ This existential concept was introduced in Chapter Three (see: *Freedom and Responsibility*).

¹⁹⁷ My use of the term 'man' is a deliberate discursive tactic here.

FINAL CHAPTER: WHERE TO FROM HERE FOR GENETIC COUNSELLING?

TROUBLING THE 'PROMISE' OF TECHNOLOGY

Due to the availability of genetic testing technologies, the parents in this study seem to see the state of being-a-carrier as more determinate than it is; the decisions ahead of children affected by this genetic threat more clear-cut than they can be. It could be said that a positive carrier test for a common mutation in the CF gene is determinate in the sense that one then knows for sure that they are a carrier (a negative carrier test result only highly reduces their risk of being a carrier). However, it becomes an indeterminate state when one is unsure of the status of one's partner, when one appreciates the risks involved in conceiving, when one has mixed feelings about the option of terminating a pregnancy, when the prognosis for the disease is becoming more equivocal (although for the parents and CF sufferers there is much hard work to be done for many years just the same), and when living in a society where there are diverse views around responsibilities regarding pregnancy and parenthood. Now too, more and more mutations in the CF gene are being discovered; some deleterious, some seemingly harmless polymorphisms, some ambiguous (how can we be sure when the latter kind will 'cause' CF and when they will not?).

Technology was ever-present in the interviews with the participants in this study. In this thesis, I have juxtaposed the faith, hope and trust that parents have in relation to the certain improvements they believe Technology will bring as against its limitations, its risks, and its potential to close off possibilities. Only those who have lost a sibling to CF do not operate on hope. They know otherwise. While those who face genetic threat see that Technology has improved to date and, with no evidence to the contrary, believe that it will continue to do so, the CF 'survivors' have lived through the suffering inflicted when Technology falls out of step with hope.

It has been said that an “imaginative re-vision [is] required so that we consider not just ‘where in the world’ we are going with the new genetics ¹⁹⁸, but where we want to go and whether we in fact want genetics to lead us there” (Lippman, 1991, p.49). We must trouble the notion that Technology will eliminate suffering. We must trouble the notion that Technology will eliminate risk. We have seen that there is suffering to be had, risk to be managed, and responsibility to be considered in decisions made in the genetic counselling/testing context. The choices ahead of the children who we know as ‘CF carriers’ will not necessarily be ones made from a situation of *Freedom*, and the dilemmas they face are not easily resolved.

UNDERSTANDING THAT GENETICS ‘PRODUCES’ IDENTITY

The claim that it is in DNA that science could discover the source of our sameness and the root of our difference not only points to the central vision of modern genetics (that it has at its disposal the means for mapping human identity and difference) but also to why genetics is of ethical interest. (Diprose, 2005, p.237)¹⁹⁹

Novas and Rose (2000, p.506) have said that the genetic counselling session is now valued “not so much for reproductive advice, but in terms of providing the coping skills necessary for coming to know the truth about one’s genetic self”. But what is this ‘truth’?

Featherstone, Latimer, Atkinson, Pilz, and Clarke (2005, p.553) describe the “spectacular presentation and representation of patients, their bodies and their identities within the clinical space defined by contemporary genetic medicine”. They claim to trace a broad historical and cultural pattern “that brings together the

¹⁹⁸ Here, Lippman cites Fletcher (1989).

¹⁹⁹ In saying this, Diprose is alluding to a quote from Weldon (1989).

*spectacular display*²⁰⁰ and the *oracular pronouncement*²⁰¹ as long-standing (although by no means immutable) features of medical knowledge" (2005, p.553). Their discussion shows the potential for identities to "[be] re-defined by molecular tests and re-negotiated by the clinic" (2005, p.568). The genetics laboratory and genetic counselling consultation are now also sites for the enactment of "oracular authority"²⁰² by genetic scientists and clinical geneticists. While there is no dysmorphology associated with being a carrier, the clinicians are tenacious; their gaze extending ever further – contemporary genetic medicine gives them unprecedented access to the previously **unseen**; to our **true** identity.

Unlike the patients who are the object of the dysmorphologist's clinical gaze (as discussed by Featherstone et al., 2005, p.565-566), gene carriers have no "look". But "[d]iagnosis and clinical classification are being reshaped by genetic technologies" (Featherstone et al., 2005, p.567) as our trust in them becomes more complete – the well can now be diagnosed and classified too. While their 'diagnosis' is beyond the reach of the clinician, it is not beyond the reach of the geneticist. The monstrous DNA²⁰³ is triumphantly revealed – a diagnosis is within reach but it is only accessible to the '**penetrating gaze**'²⁰⁴ of genetic testing technologies. The oracular pronouncement once again rings out in the clinical

²⁰⁰ In their paper looking at the features of clinical dysmorphology and its processes of classification, Featherstone *et al.* describe the "adjudication of dysmorphology [as] a contemporary exemplar of the spectacular" (2005, p.551). They say that "visual display is a long-standing feature of medical knowledge" and emphasise the "the role of visual representation in the creation and transmission of medical knowledge" (2005, p.555).

²⁰¹ This term refers to the display of clinical authority "through the narration of professional 'experience' and the ability to see and de-code the signs of diseases" when assessing patients with dysmorphic features (Featherstone et al., 2005, p.562). Featherstone *et al.* found that this authority is rehearsed in clinical consultations, in which "professional authority and status [are displayed] through a number of rhetorical devices" (2005, p.562).

²⁰² I borrow this term from Featherstone *et al.* (2005). They say that, notwithstanding the emergence of genetic technologies, the ability of the clinician to "'see' a syndrome" and the clinician's "warrant of personal knowledge" continue to enact authority in the clinic (2005, p.564/571). In fact, they found that "status is enhanced with colleagues by the minimal use of technologies to make a classification" (2005, p.565).

²⁰³ Remember my extrapolation of Rosi Braidotti's teratology work here (see section in Chapter Six: Concluding Remarks).

²⁰⁴ This is my own term. I prefer it to the 'molecular gaze' of Novas and Rose (2000, p.487) because it denotes something of the **desire** of the seer and the knowledge **incarnate** that it seeks.

space: 'You **may** have a child with cystic fibrosis. **Carry** this prophetic knowledge with you for you now know the **truth** about yourself!'

Featherstone *et al.* say "[t]he work of the genetics service includes the ascription of specific named conditions to patients" (2005, p.571). The need to assign an identity to the client (i.e., to name them²⁰⁵) despite lack of pathology or dysmorphology could be said to be a relic of the clinic's "symbolic and functional power: the gaze of the clinician [...] still exerts [its] influence" (Featherstone *et al.*, 2005, p.571). It still sees – only this time it is monstrous DNA that is seen, rather than a monstrous body. The 'carrier' is a continuation of "the spectacle of the clinic" ... with a different kind of slide show. In these slides, there are no faces; no bodies – these are superfluous to diagnostic requirements. Only a chromosome or a gene sequence is necessary in assigning an identity here. The faith in Technology is absolute, and unhindered by deference to clinical judgement. This is the 'truth' of which Novas and Rose were speaking (2000, p.506). If there is a kind of reverence associated with the ability to make oracular pronouncements based on the spectacular displays in the clinic – for Featherstone *et al.* (2005, p.553) say that "[o]racular pronouncements invoke the almost sacred gaze of the clinic" – what then of the oracle who reveals the spectacular display that cannot be seen?

Diprose explains that "[t]he point at which genetics, ethics, and poststructural critiques of models of self-present identity and difference come together is over the issue of the body and sense (meaning)" (2005, p.237). She claims that

it is as bodies that we make sense. It is as bodies that our finitude and uniqueness are signified to others; hence it is as bodies that we are both social and moral beings. This uniqueness is expressed through, and is inseparable from, being open to others within a social context of discourses (scientific, ethical, sociological). As such, identity, and therefore difference, is never self-present; the

²⁰⁵ I have previously given a presentation on "the unchallenged use of the word 'carrier' as a label for those of us who know they have a particular genetic anomaly" (Attard, 2001, p.140). I traced the etymology of the words 'carry' and 'carrier' in order "to uncover the meanings that have become deeply embedded in their use over time" and considered what impact these may have on how 'the carrier' is perceived by themselves and others in light of this (2001, p.140).

body makes sense, but never completely or in and of itself. (2005, p.237)

Diprose goes on to say that, in this schema, the true ethics of genetics

lies in recognizing the way that scientific and other discourses make sense of bodies and in ensuring that in this social expression of bodies scientific and other discourses remain open to difference and hence open to the openness by which bodies make sense (2005, p.238).

In other words, if we are to be ethical about the way we practice our 'genetics' – the way we generate our theory and apply it – we will look to understand how we are involved in 'identifying' bodies and potentially complicit in shutting out other ways of looking at them and allowing them 'to be'.

Diprose (2005, p.240) cites the work of Schenck (1986) who is inspired by Merleau-Ponty's work on our embodied being (i.e., that it is through our bodies we comport ourselves toward the world). She summarises Schenck's argument: "our body is not just an instrument by which we express ourselves" but, rather, "*is literally ourselves expressed*"²⁰⁶ (2005, p.240). With this being the case, we understand how communications that 'geneticise' the body can very readily shape who we think we are. Whereas previously it has been claimed that the processes employed within a genetic counselling consultation reveal an identity that 'is', rather than spoil an identity that 'was-not' (Armstrong et al., 1998), in this thesis I have contended that these processes (in addition to genetics theory) actually work to produce an identity that 'does-not-have-to-be' (see section in Chapter Five: Concluding Remarks).

According to Diprose, biomedical science is comfortable in its "[claim] to know, at least potentially, the identity of and difference among bodies" and "[acknowledges] a role in the observation and manipulation of that identity and difference" but it would not "confess to any constitutive role in the expression of the body's unique sense" (2005, p.243), as I am claiming here. Diprose too has insisted that biomedical science "has a role in the expression of our being-in-the-world and is not

²⁰⁶ Diprose attributes this last phrase (and its emphasis) to Schenck (1986).

just a mode of reparation of that being”, saying that “[i]t is as a discourse as much as in its practice that biomedicine is ethical” (2005, p.243).

Rather than **uncover** the difference between carriers and non-carriers, genetics actually serves to **create** that difference. This is consistent with Hegelian philosophy which, Diprose points out, reminds us that “identity is always *produced* through differential relations” (2005, p.247). Diprose (2005, p.247) concludes that the focus of genetics on “the origin of difference”, and its “[claim] to know” what this is, actually serve to produce – that is, become constitutive of – this difference. She says “genetic theory is itself a genetic operation; it is involved in the production of identity and difference”, and warns that, as such, it “runs the risk [...] of effecting an ontological closure to otherness” (2005, p.247). In line with this, and the findings of this study, **the operation of genetics can be seen to bring ‘the carrier’ into existence, genetically**. This assumed identity is mutually constituted between ‘genetics’ (its theory and practice) and the individual concerned, depending on how the genetic information comes to be viewed; comes to be known (see section in Chapter Five: The Gene and Its Relationship to Identity). Genetics would see that it brings to light the hidden, but already-existing, truth about “the source of our sameness and the root of our difference” but it actually plays a part in creating ‘truths’. And naming them.

‘The carrier’ now exists.

Yet there is room to conceptualise it as still more than a productive label. Donna Haraway would say that we could describe this thing we call ‘carrier’ as a “frozen story” (Schneider, 2005, p.127). In an interview with Schneider, Haraway talks about a kind of “story-telling practice” – one in which you take an object, “unpack” it and become “connected” to the world histories that comprise it (cited in Schneider, 2005, p.127-128). She demonstrates how that object then becomes “[something akin to] a metaphor very fast” – or, at least, “little condensations that you sense are tiny intense knots of something much bigger” (cited in Schneider, 2005, p.128). In Haraway’s schema, then, ‘the carrier’ would be described as a

literal metaphor that allows us to see the biological and social and technological and existential histories that comprise it. It is indeed a little condensation that is a tiny intense knot of something much bigger – the existential dilemmas that come along with being human. While we acknowledge there is a simple empirical description of what the carrier state is, in this thesis we have seen that it is also a metaphorical object that superbly fulfils its “[threat] to explode into universes of meaning” (Haraway, cited in Schneider, 2005, p.128).

MAKING DECISIONS ACCORDING TO THE EXISTENTIALIST ETHIC

Diprose describes ethics, in general terms, as “the question of being positioned and taking up a position in relation to others” (2005, p.238). In this account, I have shown that existentialist philosophy can inform an ethic for genetic service providers. The existential concepts of *Freedom* and *Authenticity* are at the heart of the existentialist ethic, which has been delineated here. It was also possible to position Feinberg’s child’s “right to an open future” as an extension of this because it has strong resonance with the existentialists’ call that we actively work to protect and expand the freedom of others. Introducing the existentialist ethic into the ‘genetic testing of children’ arena offers a guide to decision-making for parents and health/genetics professionals. It, teamed with Feinberg’s “child’s right to an open future”, can assist by offering a flexible approach to working through real-life scenarios and concrete ethical dilemmas in the genetic counselling/testing context. I suggest that it may be useful in other aspects of health care delivery also.

Providing genetic counselling in a way that “empowers the person, fosters their autonomy, and evokes their competency” can demand a great deal from the genetics professional (S. Kessler, 1997, p.293). Furthermore, S. Kessler (1997, p.293) says these aspects of genetic counselling can be “limited in application” and “difficult to use” due to circumstances beyond the control of the individual genetic counsellor (e.g., a health care system “in which efficiency, profit, or other factors rather than the needs of clients dictate what transpires in professional-client interactions”). Despite this, Kessler remains optimistic that “**creative professionals**

will find ways, as they always have, to retain the human side of their work” (1997, p.293). I believe that the existentialist ethic can assist the individual genetic counsellor to do this. While its underlying philosophy is a difficult read for the non-philosopher, the overall crux of it is readily understood, accessible and practicable: **maintain a focus on *Freedom!*** And in the case of children: **think “right to an open future”**.

Holm has said that a health care ethic must not just “show us an ideal picture of how we ought to be as people, but it [must also] tell us how we can attain this ideal state” (2001, p.31). He went on to say that

[p]art of the function of a health care ethics is to be a basis for education in the health care professions, in order to ‘produce’ professionals of a high ethical standard. It is therefore not sufficient to have an ethical framework that informs us about the final product, the expert ethical professional, unless the framework also contains resources that enable us to develop methods to ‘manufacture’ this product. (2001, p.31)

I concur with Holm on this point. If the existentialist ethic is to be used to develop an ethical framework for genetic counselling services, then it must also facilitate the creation of the “expert ethical professional”. To this end, there is a plethora of existentialist texts that can be ‘workshopped’ in classes with students and professionals. The overview I have provided here of the philosophy, my explication of its central concepts and my delineation of the existentialist ethic provide a further, perhaps more readily understandable, resource for the non-philosopher.

Crucial to the process of ‘learning’ the existentialist ethic, however, will be the attitude of the student. Simone de Beauvoir offers an insight here:

A book is a collective object. Readers contribute as much as the author to its creation; and mine, like myself, were concerned with morality; I had adopted a perspective so natural to them that they took what it showed them for reality itself. Beneath the veneer of abstract concepts and edifying sentences, they perceived the emotion so clumsily buried there; they brought it back to life; it

was their own blood and their own life that they were lending to my characters. (1965, p.38)

Citing Umberto Eco (1992), Crotty (1998, p.105-106) too has drawn attention to the importance of the “dialectical link” between “*intentio operis* (literally, the ‘intention of the work’, that is, the purpose expressed in and by the text itself) over against the *intentio lectoris* (the ‘intention of the reader’, that is, the personal purpose that the reader brings to the reading or infuses into the reading)”. On this basis, we see that the creation of the expert (existentialist) ethical professional will rely upon the student being willing to fully engage with the texts in order to co-create with the author a ‘working ethic’ that makes sense to them as individuals. The health professional who wishes to deepen their ethical orientation to their practice will need to reflect upon the dual intentions present when reading existential works. In this way, they can integrate its ethic into the way they reflect upon, and go about, their work.

FOSTERING EXISTENTIAL INDIVIDUALITY IN THE GENETIC COUNSELLING/TESTING CONTEXT

Can ‘the carrier’ make a free choice in the genetic counselling/testing context? How *free* can we expect any “embodied and socially situated agent” to be (Flynn, 2006, p.50)? In his discussion of *Freedom*, Merleau-Ponty said that Scheler (n.d.) countered the Kantian idea that an intention is tantamount to the act with an argument along the following lines:

[T]he cripple who would like to be able to save a drowning man and the good swimmer who actually saves him do not have the same experience of autonomy. The very idea of choice vanishes, for to choose is to choose *something* in which freedom sees, at least for a moment, a symbol of itself. There is free choice only if freedom comes into play in its decision, and posits the situation chosen as a situation of freedom. (Merleau-Ponty, 1962, p.437)

In this thesis, I have drawn attention to choices made by participants that may at first glance have appeared to be freely made yet could hardly be said to have been so when, to their mind, in their situation, they could not have chosen other than

they did. These were decisions made in which *Freedom* did not come into play and the situation chosen was not one of *Freedom*. Commonly, it was *Responsibility* towards another that tended to confine the choices a person felt they had available to them.

The Other is inevitably present in the genetic counselling/testing context. This thesis gives an insight into when it can emerge, how it manifests itself and the various forms it can take – e.g., the asking of how a child comes to be born with CF (see section in Chapter Five: Shame and Stigma Associated With Having ‘Something Wrong’), “a lot of the nurses and a lot of the other mums” (see section in Chapter Five: The Influence of Others), “an older doctor”, genetic counsellors who “just listened” (see section in Chapter Five: Josie’s Tale: “Oh, Here I Go Getting Another Blasting!”), an existing child (see section in Chapter Five: When Termination Is Not An Option), a yet-to-be-conceived child (see section in Chapter Five: When Having Another Child Is Not An Option), parents who resist “prejudging the information” (see section in Chapter Five: Protected Rights of the Child), parents who hope their child will “do the right thing and be tested” (see section in Chapter Seven: Onus to ‘Use’ the Technology).

We have seen that the Other does influence how an individual evaluates genetic information and is given consideration in the context of the decisions they make in relation to it. Their possibilities are impinged upon by the possibilities of others. They can never control what other people think of them, their gene defect and their choices. They see themselves in the eyes of others and it takes work to ensure they do not concede that feelings generated by *The Look* are the final word on who they think they are. It takes work to escape aspects of their self-image that are wholly shaped by the judgement of others; to overcome that *Self-estrangement* or that gap between how they would like to act and how they feel they must act, that would not exist if what no one else thought mattered. Given this, the challenges to the individual’s ability to make *free*, autonomous decisions are significant.

The ASGC Code of Ethics (2008, p.1-2) says:

Genetic counsellors are personally accountable for their practice and conduct, which must respect the reasonable interests of individual clients, aim to enhance the general health and well-being of the community, justify public trust, and enhance the reputation of the profession.

Genetic counsellors must be interested in the ability of their clients to make autonomous decisions, as far as possible, from a position of immersion in the 'they'. Indeed, it has been claimed that respect for client autonomy is greater in the profession of genetic counselling than in almost any other area of medicine (Davis, 2001, p.12). This may be so but Clarke has previously contended that claims to non-directiveness by genetic counsellors could be seen as disingenuous given that little investigation has taken place into whether clients are indeed able to achieve true autonomy within the genetic testing and reproductive decision-making contexts (1998, p.5). He wrote:

Insufficient attention has been paid to social influences either at the macro level (e.g., social tolerance; the political willingness to support those with genetic disorders and other types of disability) or at the micro level (e.g., the way in which genetic information or testing is made available in routine clinical practice; the way in which family, friends and neighbours respond when a genetic condition is identified; personal experiences of stigmatization or discrimination) (Clarke, 1998, p.5).

This study has paid attention to the accounts given by parents in 'carrier families' about their experiences and it has looked at what this can tell us about the social influences that are operating about them at both a macro and a micro level.

Heidegger's writing on 'thrownness'²⁰⁷ could provide for an interesting account of how the differing contexts of *Dasein* (i.e., historical, social, etc.) contribute to the variation in the human response to our geneticised world. If *Dasein* makes sense of itself out of the world into which it is thrown, then it could be said that *Dasein* makes sense of its own carrier status out of the world into which it is thrown. In

²⁰⁷ The meaning of this term was discussed in Chapter Three (see section: *Freedom and Choice*).

this world, Heidegger says *Dasein* is dominated by others; “taken over unawares” by them, and that this serves to entrench their power (Heidegger, 1996, p.118).

Continuing exploration of how *Dasein* is dominated by others in the genetic counselling/testing context – how the ‘they’ takes over *Dasein*’s experience of the carrier state unawares and how the ‘they’ shapes *Dasein*’s behaviour in relation to genetic information – is required. What kind of power, and whose, is being entrenched here?

I now draw a parallel between achieving autonomy in the genetic counselling/testing context and the achievement of existential individuality in life more generally. Existentialists are troubled by the “social and economic pressures of mass society for superficiality and conformism” and the ability of the individual to live *authentically* amidst this (Flynn, 2006, p.8). Similarly, we must be troubled by, and trouble, the notion that the individual is able to achieve autonomy in today’s geneticised and technologised realm of reproductive decision-making. If, as human individuals, we are, and have always been, entangled in the lives of others as Løgstrup said (Holm, 2001, p.27-28), how can we know where our selfhood begins and the values of those close to us (and even far from us) end? Flynn said that “[t]o be truly authentic is to have realized one’s individuality and vice versa”, continuing that “[b]oth existential ‘individuality’ and ‘authenticity’ are achievement words” (2006, p.74). How can we possibly know when a decision from a position of autonomy has been achieved? Can a genetic counsellor ever strike this holy grail of the profession? In what setting will they find it? Who can tell them how to get there? And how will they know it when they see it?

Notwithstanding the difficulty in attaining this goal in its purest form, it is hoped that this thesis will be seen as having made a contribution toward the fostering of existential individuality in the genetic counselling/testing context. By holding up the ideals of autonomy/*Authenticity*, one presumes that, at the very least, counselees will be supported to make a decision which seems most appropriate to them in their ‘situation’. To do this may require that genetic counsellors ensure their clients recognise any external pressures that may be subtly influencing their decision-

making. It may entail striving to reduce the impact that the perceived expectations of society have on their decisions. It may see genetic counsellors become part of a proactive push to increase respect for people with genetic disorders and disabilities generally, and to lower stigmatisation and discrimination attached to these conditions²⁰⁸. This is if they wish their claims that clients are able to make their own autonomous decisions in a 'value-neutral, even-handed, impartial and non-coercive'²⁰⁹ environment to be taken seriously.

SAFEGUARDING THE CHILD'S RIGHT TO AN OPEN FUTURE

As mentioned in Chapter One (see section: Ethical Issues Facing Genetic Counsellors: Dealing With Past, Present and Future), confidentiality and privacy concerns in genetics do focus on employment and insurance issues but there are also important privacy concerns in families. Davis (2001, p.85) says that "[o]ur society, our legal system and the medical profession give great deference to the privacy of reproductive decisions"... but can we say that parents and family members always do the same? As we have seen in this study, it may sometimes be family members who are the greatest threat to the autonomy of the individual making decisions.

Davis believes that "the autonomy of the individual is ethically prior to the autonomy of the family [in which they were raised], even though, chronologically and developmentally, it is certainly the other way around where children are concerned" (2001, p.31). While Davis agrees the liberal state must support parental autonomy, she points to where the line must be drawn with this, saying that parents who wish to exercise their right to bring up their children according to their sense of what is "good" must not do so in a way that "threatens to extinguish the

²⁰⁸ Lippman has previously commented that "[s]ociety does not truly accept children with disabilities or provide assistance for their nurturance", concluding that, "[t]hus, a woman may see no realistic alternative to diagnosing and aborting a fetus likely to be affected" (1991, p.32).

²⁰⁹ We replaced the pursuit of non-directiveness with these principles in Chapter One (see section: Is Non-Directive Genetic Counselling Possible?).

abilities of children to choose their own lives when they become adults" (2001, p.31):

[M]orally the child is first and foremost an end in herself. Good parenthood requires a balance between having a child for our own sakes and being open to the moral reality that the child will exist for *her* own sake, with her own talents and weaknesses, propensities and interests, and with her own life to make. [...] Parental practices that close exits virtually forever are insufficiently attentive to the child as an end in herself. By closing off the child's right to an open future, they define the child as an entity who exists to fulfill parental hopes and dreams, not her own. (Davis, 2001, p.34)

While we must "strive to shape the values and lives of the children in [our] care", that shaping must not "[impinge] substantially and irrevocably on the child's right to an open future" (Davis, 2001, p.32). The difficulty in applying this ethical 'line' to the question of the carrier testing of children is deciding whether it **substantially** restricts the children's rights and, because they may still choose not to have their future partner carrier-tested or not to have prenatal testing, making a call as to whether it is **irrevocable** (although they will still need to make these choices in relation to having the definite knowledge that they are carriers).

Davis argues that "the extreme respect for the autonomy of the parents-to-be, who are the obvious clients of geneticists, has sometimes obscured concern for the autonomy of the child-to-be, who is in an important way also the geneticist's client, or at least the object of her concern" (Davis, 2001, p.6-7). The rights of the adult who the already-born or yet-to-be-born child is-to-become are particularly compelling in light of the attention I have given in this thesis to the existentialist ethic and the child's right to an open future. These demand that the rights-in-trust of children, who are equally the genetic counsellor's 'clients', be safeguarded. The responsibility of the genetic counsellor becomes clearer when the already-born and the yet-to-be-born are seen as clients, alongside their parents.

Davis concludes her excellent work *Genetic Dilemmas: Reproductive Technology, Parental Choices, and Children's Futures* by saying that “we should use these new technologies to ensure for our children, and for their children, not more constricted futures but more open ones” (2001, p.131). I agree. Legislators must support this. Policy makers must support this. Health professionals must support this. Genetic counsellors must support this. And, very importantly, parents need to support this for their children too. In this thesis, we have seen that Technology can be constricting (e.g., see section in Chapter Seven: Onus to ‘Use’ the Technology) but also that Technology can co-exist with an open future (e.g., see section in Chapter Seven: Sanctioning the Choice of Inaction). Those listed above all have some part to play in determining which of these occurs. I echo Davis’ call (2001, p.85) that “more research is needed on the appropriate age at which to undertake testing and screening for genetic disorders” and agree that this should be judged against expanding the possibilities of those tested rather than limiting them.

POSITIONING PARENTS AS CARRIERS OF RESPONSIBILITY

As seen in this thesis, there are parents who see the issue of when and how to tell their children they do, or may, carry a recessive disease allele as one riddled with dilemma (e.g., see section in Chapter Six: Waiting for the Right Time **and** section in Chapter Five: Protected Rights of the Child). My feel from the interviews with the parents in this study is that many would have been receptive to advice and information about what is thought to be the best way to ‘care for’ this genetic information (if it were available). I think parents will be more readily able to put recommendations of this kind into practice if they are based on empirical evidence that relates to an understanding of children’s well-being, rather than on an attempt to order the (sometimes conflicting) values within a principle-based ethics.

In the context of genetic information, my firm recommendation is that a child’s well-being be framed as inexorably linked with their right to an open future (this also resonating with the existentialist ethic). Wherever possible this will mean the avoidance of determining information about a child’s genotype with respect to

disease gene alleles. And when genetic information does become known, I do not see that it is enough that the custodians are 'educated' as to the mode of inheritance and the nature of the risk it brings to its carrier, and 'counselled' with regard to managing this genetic threat. It is my contention that the custodians of that information must also be sensitised to the position of trust and responsibility they take up in relation to it when it pertains to a child. I suggest this feature prominently in the genetic counselling consultation, alongside helping parents to "understand and adapt"²¹⁰ to the information. They themselves may carry a gene but they also now carry a responsibility to 'take care' of the genetic information they have about their child. Both aspects of parenting-a-carrier need to be put to the custodians: with the genetic **information** comes **responsibility**. And, in this particular context, I am not talking about the 'genetic responsibility' that has been discussed elsewhere (Arribas-Ayllon, Sarangi, & Clarke, 2008; Novas & Rose, 2000) but, quite simply, parental responsibility. In this way, the non-carrier parent is a 'carrier' too – a carrier of responsibility with respect to their child's genetic information, with an obligation to care for it and their child's right to an open future accordingly.

When all is said and done, and even in our geneticised world, **we see that this ethical issue is as much about parenting as it is genetics**. This is good news (to some extent). At a micro level, we see that parents still have an opportunity to safeguard as far as possible their child's right to an open future. And genetic counsellors can support this process. But there is a limit to what they can achieve. Enactment of parental responsibility will only 'reach' so far. This thesis has shown that there is potential for the geneticisation and technologisation that is occurring at a societal level to restrict the possibilities of individuals – children and adults alike. At a macro level, the process of geneticisation is continuing to make the social world that children are growing up in today more and more focussed on their

²¹⁰ In Chapter One (see section: What is Genetic Counselling?), I made reference to the code of ethics for genetic counsellors in Australia that defines their practice as "a communication process, which aims to help individuals, couples and families **understand and adapt** to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions" ("Australasian Society of Genetic Counsellors (ASGC) Code of Ethics," 2008, p.1).

genes. Is it inevitable – even unstoppable – that this will continue unabated? Perhaps parental responsibility will manifest as a collective action at a macro level. How can one salvage *Freedom* for oneself and all others in the face of this? And who is going to take *Responsibility* for doing it?

CONCLUSION: WHAT HAS BEEN ACHIEVED HERE?

As has been stated, my broad aim at the outset of this project was to make an ethical and knowledge-based contribution to the debate about the genetic testing of children. The purpose of this interpretive study was to provide health professionals and genetic counsellors with an insight into the experiences of ‘carrier families’ in order that this impact positively on child and family well-being in those families who come into contact with genetic counselling services in the future.

The individual stories told here are specifically about families affected by the threat of a mutation in the cystic fibrosis gene. The findings of this study are not totally transferable but they are likely to be highly relevant to other families who come into contact with genetic counselling services, particularly those also living-with the ‘threat’ of a recessive disease allele. My findings will not give genetic counsellors the ability to pre-empt and therefore emotionally regulate their next counsellee’s reaction to this and similar genetic threats. However, they will give genetics professionals an **idea** of how a particular result may make a person feel, an **insight** into the issues that may need to be addressed, and the **illumination** required for achieving empathy and tact in their practice. This is the kind of knowledge-based contribution to the betterment of the practice of genetic counselling that Nancy Wexler (1979) first called for thirty years ago (see section in Chapter One: A Call to Research!).

My thinking has been guided by an appeal to existential philosophy, which highlights the ambiguities and the tensions within human existence; its dilemmas and its paradoxes. I have developed an analytical framework based on my explication of concepts from the existentialist literature. Applying this conceptual

framework to the analysis of my data has, I believe, in turn, showed that the data itself can add to an understanding of these concepts. I suggest that this resonance between the concepts and my data has validated the methodological approach that I adopted in relation to this study. This conceptual framework is now available for other researchers to apply creatively to the analysis of their data across a wide range of human experiences.

An existential analysis has allowed me to leave the ambiguities and the contradictions within and between stories unresolved²¹¹. Introducing the existentialist ethic into the 'genetic testing of children' arena has offered a novel approach to the examination of ethical dilemmas that emerge there. I have delineated this ethic and shown that it provides a means for us to pass judgement in relation to the choices of others in the genetic counselling/testing context, as against their adherence to the values of existentialist thought. It has allowed us to examine which stated attitudes and actions are working to expand or to limit the future possibilities of children. The rights-in-trust of the already-born or yet-to-be-born child are particularly compelling in light of the attention I have given in this thesis to the existentialist ethic and the child's right to an open future. We feel hope, even from within a geneticised world, when we see the pedagogic thoughtfulness-and-tact-in-action of parents who are properly governed by a sensitivity to these rights.

I hope that I have also been able to relate something of interest here about the emergence of a laboratory-trained geneticist from a position of immersion in the 'they' world of science and positivist approaches to research; from the geneticised ways she had come to understand people and their possibilities. It has not been my intention that those who come into company with this thesis will part ways with it

²¹¹ This aspect of the study, at least, would receive the approval of Donna Haraway. Her writing, "quite intentionally", does not "resolve into theoretical or even narrative wholes; it does not aim to 'tie up' loose threads into a 'whole cloth' of orderliness either of feminism or science studies" (Schneider, 2005, p.157). Haraway welcomes contradiction – "she forefronts it both as a productive way to think and imagine and because she insists that it is so easy to see in what she studies. She mixes things that are not usually mixed and asks for multiple and often diverse literacies in her readers" (Schneider, 2005, p.158).

believing that it makes claim to a definitive statement about the beneficence of genetic testing (or otherwise) or a definitive pronouncement that there are inevitabilities attached to the carrier testing of children. But it is my intention that it be seen to be insisting that we talk about genetic testing for what it is; that we see the issue of the carrier testing of children in all its reality.

Carrier testing is one way of dealing with a particular problem – a problem that is about much more than genetics. The problem of reproductive ‘success’ (that there are failures too is inferred) is also culturally-bound, historically-bound, sexually-bound, gender-bound, racially-bound, politically-bound, economically-bound and, of course, existentially-bound. When we remember this, it becomes clear that the geneticisation of this problem cannot be **the** solution to it. How could it be? And in the forgetting of this, we see that there could be many repercussions of which we can only begin to hazard a guess.

At the very least, we must talk about genetic testing for what it is – a procedure that gives a person some information about their genetic make-up, which may or may not influence their decision as to whether they will have children and/or their decision as to which of their embryos- or foetuses-in-waiting they will implant or give birth to. It will not entirely prevent suffering. And sometimes it will be the cause of it. **Choose your pain.**

GLOSSARY OF TERMS

- Absurdity*** The existential concept of the *Absurd* recognises that, despite the human longing for it not to be so, our lives are ultimately without meaning and the Universe is indifferent to this. We rise above our *absurd* fate “not by dull resignation but by *deliberate choice*” (Flynn, 2006, p.48).
- Ambiguity*** According to existentialists, there are many tensions in human existence which render it *ambiguous*. To exist authentically we must face up to this *Ambiguity*, recognising the many poles between which our existence stands and allowing each to assert itself.
- Angst*** *Existential Angst* refers to the emotion we feel when we experience an awareness of our *Freedom*.
- Authenticity*** When a person reaches a point where they can develop their own value system (their own set of ethics) in accordance with their own beliefs, and use this to interpret their own situation and guide the way that they behave, then existentialists would say they are now living an *authentic* existence.
- Bad Faith*** The existential concept of *Bad Faith* is “as paradoxical as consciousness itself. [...] [B]ad faith is ‘knowledge that is ignorant and ignorance that knows better’²¹². And this occurs within the unity of one and the same implicit self-consciousness” (Flynn, 2006, p.73).

²¹² Here, Flynn cites Sartre (n.d.).

- Being** 'Being' (always capitalised) is the term Heidegger used to denote the basic condition of our existence; it is "that primordial condition or 'ground' which allows [beings] to come into existence" (Lemay & Pitts, 1994, p.33-34).
- Care** Heidegger believed the human condition is defined by *Care*, the alternative to the *Technological Attitude*. The concept of *Care* expresses our fundamental relation to the world in which we live, "whose contents are articulated in terms of the significance they have through the intentional projects in which we engage" (Cooper, 1990, p.157).
- Dasein** A Heideggerian term which "denotes 'the manner of Being' possessed by creatures like ourselves" (i.e., self-conscious creatures) (Cooper, 1990, p.67). It is synonymous with Sartre's '*être-pour-soi*' (Being-for-itself) and Merleau-Ponty's 'man-in-the-world' (Charlesworth, 1975, p.24).
- Despair** The existential concept of *Despair* does not refer to a particular emotional state but rather to the feeling that comes with the knowledge that we only have a limited part to play in shaping events that happen in the world around us.
- Eugenic** The practice of (forcibly) subordinating or sacrificing "the rights and welfare of individuals [...] to the supposed interests of collective entities such as society, the state, the race or the species" (Clarke, 1998, p.1).
- Freedom** *Freedom* is commonly acknowledged as the supreme value of existentialist thought. Refer Chapter Three for an overview (see sections: *Freedom and Responsibility* and *Freedom and Choice*).

(The) Look

The Look, as an existential concept, refers to the phenomenon whereby a person becomes acutely conscious of how they appear in their objectification before the Other. *The Look* reveals to us that it is in the eyes of others that we discover ourselves.

Nausea

The existential concept of *Nausea* describes the feeling of malaise and melancholia that overwhelms a person when confronted by their own contingent existence – their own superfluity – indeed, by the contingent and superfluous nature of everything around them.

Predominance of the Other

Self-estrangement through the *Predominance of the Other* is a particular form of *Bad Faith* where a person assumes an identity and behaves in a way that too completely conforms with who other people expect them to be, putting aside their own innermost aspirations as to the kind of life they would like to lead.

Responsibility

Existential Freedom brings with it concomitant *Responsibility*. Refer Chapter Three for an overview (see section: *Freedom and Responsibility*).

Revolt

It is through recognising and accepting our *absurd* fate that we transcend our fate. To “[forge] ahead like Sisyphus despite the presumed indifference of the Universe” constitutes the supreme act of *Existential Revolt* (Flynn, 2006, p.54).

Sartrean Anguish This existential concept encapsulates “the experience we have when we realise our total responsibility for ourselves” (Charlesworth, 1975, p.9).

Sartrean Fear This existential concept denotes “the experience we have when we contemplate being caught in the deterministic process of nature” (Charlesworth, 1975, p.9).

Self-estrangement The existential concept of *Self-estrangement* is probably best understood as a term used to describe the existence of a person who is making decisions and living their life according to the wishes and values of others. They are said to have become alienated from themselves.

Technological Attitude In the *Technological Attitude*, we position ourselves as the ‘thinking thing’ against all other beings, which are seen as something there for us to consume; as ‘stuff’ which exists exclusively for our purposes.

(The) ‘they’ Immersion in the ‘they’ constitutes loss of self. This excerpt from Karl Jaspers’ work describes how this occurs:

“The sort and extent of my being as a social I is stamped upon me in the ineradicable concatenation of my life with human society. Thus conditioned to be a historic particular, an existence of my world, I am what everybody takes me to be in this context. In rationalized societies the very substantiality of the particular will be more and more whittled down, until in borderline cases my belief in the historic meaning of the whole in this state will extinguish all sense of substance in my specific existence. Left to me, then, is nothing but the social existence in which I am whatever rights and duties I am given. Everyone is like everyone else, reduced in principle to a specimen that is to share and share alike in the social possibilities of welfare, of work, of enjoyment. What I come to be as this social I is ‘all of us.’” (1970, p.30)

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CONDITIONAL APPROVAL LETTER FROM UNIVERSITY RESEARCH ETHICS COMMITTEE

[UNIVERSITY LETTERHEAD]

*Social and Behavioural Research Ethics
Committee
Faculty of Social Sciences*

SBRE 2143

20 September, 2000

[Researcher's address]

Dear Ms Attard

Project 2143: Understanding the experiences of parents who know that their child is, or could be, a carrier of cystic fibrosis

At its meeting on 18 September, 2000 the Social and Behavioural Research Ethics Committee considered the application you submitted in respect of the above project.

The proposed project has been given conditional approval subject to:

- i) Advice as to who will identify potential participants from hospital files.
- ii) An undertaking that staff will be asked to indicate their willingness to be involved.

Please submit further information relating to, or confirmation of, the above matters to me in writing as soon as possible to enable approval of the project to be confirmed. My office is located in [Room], [Building].

May I draw to your attention that if you receive funding for your research through the National Health & Medical Research Council, it will be necessary for you to comply with special reporting requirements. If this is applicable to your research, please contact me and I will inform you of the relevant procedures.

Yours sincerely

[Name]
Secretary
SOCIAL AND BEHAVIOURAL RESEARCH ETHICS COMMITTEE

**FINAL APPROVAL LETTER FROM UNIVERSITY RESEARCH ETHICS
COMMITTEE**

[UNIVERSITY LETTERHEAD]

*Social and Behavioural Research Ethics Committee
Faculty of Social Sciences*

SBRE 2143

1 May, 2001

[Researcher's address]

Dear Ms Attard

**Project 2143: Understanding the experiences of parents who know
that their child is, or could be, a carrier of cystic fibrosis**

Further to my letter dated 20 September, 2000, I am pleased to inform you that approval of the above project has been confirmed following receipt of the additional information you submitted in April 2001.

Yours sincerely

[Name]

Secretary

SOCIAL AND BEHAVIOURAL RESEARCH ETHICS COMMITTEE

CONDITIONAL APPROVAL LETTER FROM HOSPITAL RESEARCH ETHICS COMMITTEE

[HOSPITAL LETTERHEAD]

30th October 2000

[Researcher's address]

Dear Ms Attard

re: Understanding the experiences of parents who know that their child is, or could be, a carrier of cystic fibrosis. REC1164/10/2003

Thank you for submitting the above protocol to the [Hospital Name] Research Ethics Committee. It was approved at the Committee's meeting on 25th October 2000, subject to modification to the planned selection process. The Committee considered the proposed method of subject selection might bias results and that, in any case, it was unlikely you would obtain the information you are seeking from the files. Therefore, consecutive records should be used for recruitment of subjects.

I remind you approval is given subject to:

- immediate notification of any serious or unexpected adverse events to subjects;
- immediate notification of any unforeseen events that might affect continued ethical acceptability of the project;
- submission of any proposed changes to the original protocol. Such changes must be approved by the Committee before they are implemented;
- immediate advice, giving reasons, if the protocol is discontinued before its completion;
- submission of a brief annual report on the state of progress of the study, and a final report when it is completed.

Approval is given for a period of three (3) years only, and if the study is more prolonged than this, a new submission will be required. Please note the approval number above indicates the month and year in which approval expires and it should be used in any future communication.

Yours sincerely

[Name]

CHAIR

[HOSPITAL NAME] RESEARCH ETHICS COMMITTEE

LETTER TO HOSPITAL RESEARCH ETHICS COMMITTEE

28th November 2000

Dr [Surname]
Chair, [Hospital Name] Research Ethics Committee

Dear Dr [Surname],

**Re: Research Ethics Committee's approval of my project protocol.
REC1164/10/2003**

I would like to thank the [Hospital Name] Research Ethics Committee for approving the protocol for my PhD project entitled "Understanding the experiences of parents who know that their child is, or could be, a carrier of cystic fibrosis". However, I write in regard to the Committee's request that the planned participant selection process be modified because the proposed method "might bias results" and I ask that you consider the points I make below.

I feel that perhaps I did not articulate the nature of qualitative research conducted within the interpretive paradigm adequately. Some of the concerns regarding bias in 'standard' scientific research are not compatible with qualitative research methodologies. In fact, "qualitative researchers use bias and deliberately select a biased sample" (Morse, 1998) – we seek out those who have a story to share, those who have an unusual or interesting experience to relate, and pounce on the rogues whose stories surprise us and challenge assumptions that we may take for granted!

So, qualitative sampling is geared towards finding information-rich cases, rather than towards randomisation and generalisability. Qualitative sampling does not claim 'representativeness' but, as I stated in section 2.11 of my application form, in my study "it will be important to sample for variety across the phenomenon of parenting a child who is a known or possible CF carrier". Perhaps my next comment, that "it will be necessary to identify the largest possible 'range' of potential participants" was unclear, and this is why the Committee thought I would not obtain the information I am seeking from the files. By "'range' of potential participants", I meant that I wish to interview a selection of parents who vary with regards to: 1) the ages of their children, 2) the amount of time since they found out their child is a carrier, 3) the context in which they found out their child is a carrier, 4) whether or not they have another child with CF, and, 5) their socioeconomic status. Most of this information can be gleaned from the files in the [State Name] Clinical Genetics Service and in the Dept of Chemical Pathology.

I hope this has clarified why I do not wish to use a random method of selecting participants. I have enclosed a copy of a paper by Janice Morse (1998), the editor of an international, interdisciplinary journal called *Qualitative Health Research*. She can probably explain better than me just "What's Wrong With Random Selection?"...

Yours sincerely
Melanie Attard

FINAL APPROVAL LETTER FROM HOSPITAL RESEARCH ETHICS COMMITTEE

[HOSPITAL LETTERHEAD]

12th December 2000

[Researcher's address]

Dear Ms Attard

re: Understanding the experiences of parents who know that their child is, or could be, a carrier of cystic fibrosis. REC1164/10/2003

Thank you for your letter dated 28th November 2000 in which you responded to matters raised by the [Hospital Name] Research Ethics Committee at its October meeting. All matters have been addressed and final approval is given for the study to proceed.

I remind you approval is given subject to:

- immediate notification of any serious or unexpected adverse events to subjects;
- immediate notification of any unforeseen events that might affect continued ethical acceptability of the project;
- submission of any proposed changes to the original protocol. Such changes must be approved by the Committee before they are implemented;
- immediate advice, giving reasons, if the protocol is discontinued before its completion;
- submission of a brief annual report on the state of progress of the study, and a final report when it is completed.

Approval is given for a period of three (3) years only, and if the study is more prolonged than this, a new submission will be required. Please note the approval number above indicates the month and year in which approval expires and it should be used in any future communication.

Yours sincerely

[Name]
CHAIR
[HOSPITAL NAME] RESEARCH ETHICS COMMITTEE

LETTER OF INVITATION

Dear

I am writing to invite you to participate in a research study that will document and interpret **the experiences of parents who know that their child is, or could be, a CARRIER of cystic fibrosis.**

The study is being carried out by Melanie Attard who is a PhD candidate at [Name of University] under the supervision of myself and [Supervisor B] from the School of Nursing and Midwifery. Melanie has a background in human genetics and she is trying to understand how people are affected by genetic technology. Her particular interest is carrier testing in cystic fibrosis and how it affects families.

Melanie is asking mothers and fathers of children who are known to be, or who could be, carriers of cystic fibrosis to participate in one or two informal interviews with her. She would like them to share with her what it means *to them* to know that the cystic fibrosis gene is, or could be, carried by their child. She hopes that by listening to them talk about their experiences and feelings she will be able to identify the things that are important to parents. This will add to the debate about the ethical issues surrounding genetic testing.

Melanie is being funded by an Australian Postgraduate Award from the Commonwealth Government. Her study has also been awarded funding by [her Faculty] and has been approved by research ethics committees at both the [Hospital Name] and [this University].

Participants can be assured that their contribution to the study will be highly valued and treated with utmost confidentiality.

You are under no obligation to participate in the study and if you decide not to, the treatment, care or counselling your family receives at the [Hospital Name] will not be affected in any way.

If you are willing to speak with Melanie about your family's experience of genetic testing, please read the enclosed information sheet and fill out the enclosed form indicating your willingness to be contacted. Kindly mail the 'consent to be contacted' form in the reply paid envelope prior to [date]. Melanie will then telephone you to provide more information and to answer any questions you may have. If you agree to participate, she will organise a convenient time and place to meet with you.

This research is important and your participation would be very much appreciated. Thank you for taking the time to read this letter.

Yours sincerely

[Supervisor A], Clinical Geneticist
Head, [State Name] Clinical Genetics Service

Understanding the experiences of parents who know that their child is, or could be, a carrier of cystic fibrosis.

INFORMATION SHEET

Why am I doing this study?

My name is Melanie Attard and I am a doctoral student in the School of Nursing at [University Name]. I have an Honours degree in Genetics from the University of Adelaide and I am conducting this study as the major part of my PhD project. I have undertaken this research to provide a better understanding of the issues which confront parents who know that their child is, or could be, a carrier of cystic fibrosis (CF). I hope it will draw attention to the issues that are of most importance to the parents of these children and explore what it means to parents to have this information about their child.

Participants

I would like to interview the parent(s) of children, adolescents or young adults who are known CF carriers or who have a high chance of being a carrier (i.e., if they have a brother or sister who was born with the condition, or if they have a parent who is a known CF carrier).

What is involved?

If you would like to be involved in this study, I will ask you to participate in an informal interview with me in your home or a location where you feel most comfortable. In this interview, I will ask you to share with me your thoughts related to your child's CF carrier status. Because every family's experiences are unique, these interviews will not follow a structured question-and-answer format, rather they will be 'open-ended'. This means that the 'interview' will be more like a conversation, giving you the opportunity to speak about the issues that are important to you and your family. You will be free to decline discussing any particular issue or answering any question if you do not wish to do so.

This interview will take no more than one to two hours of your time. It is possible that the outcomes of the study could be further enriched by your participation in one or two shorter follow-up interviews, however this extra commitment to the study is by all means negotiable.

With your consent, the interview(s) will be tape-recorded and later transcribed. You will then be given the opportunity to read the transcript(s), if you so wish. Please be assured that the information you provide will be treated in the strictest confidence.

Results

The information derived from these interviews will be analysed and formulated into a report that will give health professionals an important insight into what it is like to be the parent of a child who is, or has a high chance of being, a carrier of CF. It is highly likely that this research will also be relevant to families who are carriers of

other recessive conditions, and to wider issues related to the genetic testing of children. Results gained in this study will be published but neither you nor your family will be identified, and all individual information will remain confidential.

More information...

If you read this information and decide you would like to take part, I will discuss the study with you further and answer any questions you may have. If you would like to know any more at this stage, please feel free to contact me (phone: [work number] or [home number], email: [address]) or either of my supervisors, [Supervisor A] (phone: number) or [Supervisor B] (phone: [number]).

Please note that you are under no obligation to participate in the study and your decision will in no way affect the care you and your family receive at the [Hospital Name], now or in the future.

I thank you very much for taking the time to read this information!

[UNIVERSITY LETTERHEAD]

LETTER OF INTRODUCTION

Dear

This letter is to introduce Ms Melanie Attard who is a doctoral student in the School of Nursing at [Name of University]. She will produce her student card, which carries a photograph, as proof of identity.

She is undertaking research leading to the production of a thesis and other publications on the subject of: "Understanding the experiences of parents who know that their child is, or could be, a carrier of cystic fibrosis". She is conducting in-depth interviews with parents who live with this knowledge and hopes that this research will provide genetics professionals with an insight into what this means to parents and what issues are important to them.

Melanie would be most grateful if you would volunteer to spare the time to assist in this project by granting an interview which explores certain aspects of this topic. No more than one to two hours of your time would be required on this occasion. It is possible that the outcomes of the study could be further enriched by your participation in one or two shorter follow-up interviews, however this extra commitment to the study is by all means negotiable.

Be assured that any information provided will be treated in the strictest confidence and none of the participants will be individually identifiable in the resulting thesis, report or other publications. You are, of course, entirely free to discontinue your participation at any time or to decline to answer particular questions.

Since Melanie intends to make a tape recording of the interview, she will seek your consent, on the attached form, to record the interview; to use the recording or a transcription in preparing her thesis, report or other publications, on condition that your name or identity is not revealed; and to make the recording itself available to no one but us, as her two supervisors, on the same conditions. While Melanie plans at this stage to transcribe the interviews herself, time factors may render it necessary to make the recording available to secretarial assistants for transcription, in which case you may be assured that such persons will be advised of the requirement that your name or identity not be revealed and that the confidentiality of the material is respected and maintained.

Any enquiries you may have concerning this project should be directed to [Supervisor B] at the email address or telephone or fax number given above, or to [Supervisor A] (ph: [number], email: [address]). This research project has been approved by the [Name of University] Social and Behavioural Research Ethics Committee and also the [Hospital Name] Research Ethics Committee. Should you wish to discuss the approval process or voice any concern or complaint about this study, please contact the Secretary of the [University] Committee (ph: [number], email: [address]) or the Secretary of the [Hospital] Committee, [Name] (ph: [number]).

Thank you for your attention and assistance.

Yours sincerely

[Supervisor B]
School of Nursing & Midwifery
[Name of University]

[Supervisor A]
[State Name] Clinical Genetics Service
[Hospital Name]

CONSENT FORM FOR INTERVIEW(S)

I,

being over the age of 18 years, hereby consent to my involvement in the research project entitled: "Understanding the experiences of parents who know that their child is, or could be, a carrier of cystic fibrosis".

1. The nature and purpose of the research project described on the attached Information Sheet has been explained to me. I have read the Information Sheet. I understand it, and agree to taking part.
2. I acknowledge that the possible discomforts and inconveniences associated with my participation in the study, as outlined in the Information Sheet, have been explained to my satisfaction.
3. I am aware that I should retain a copy of the Information Sheet and Consent Form for future reference.
4. I agree to the researcher recording our interviews on an audio cassette. However, I understand that I may ask that the recording be stopped at any time, and that I am free to decline to answer any question.
5. I agree to the interview transcript being used by the researcher for the purposes set out in the Information Sheet.
6. I understand that:
 - I may not directly benefit from taking part in this research.
 - There will be no payment to me for taking part in this study.
 - I am free to decline taking part in the study or to withdraw from an interview session or the entire project at any time, without disadvantage. Doing so will not affect medical services or any other aspects of my own or my family's relationship with the [Hospital Name].
 - While information gained in this study may be published, I will not be identified and individual information will remain confidential.
7. I agree to the recording being made available to the researcher's supervisors.
8. I agree to the interview transcript being made available to other researchers who are not members of this research team, on condition that my identity is not revealed.
9. I have had the opportunity to discuss taking part in this research with a family member or friend.

Participant's signature.....Date.....

I certify that I have explained the study to the volunteer and consider that she/he understands what is involved and freely consents to participation.

Researcher's signature.....Date.....

10. I, the participant whose signature appears below, have been given the opportunity to view my interview transcript.

Participant's signature.....Date.....

OUTLINE OF INTERVIEW QUESTIONS

Cast your mind back and tell me about the time when you first heard of CF?

- How did you hear about it? From who? What did it mean to you at that time?

Tell me about how you first found out that the CF gene was in your family?

- How did you feel? What are your main memories about that time?

So when did you first find out that your child is a carrier?

- Is there any other situation you would compare it to?
- How would you describe the experience of that discovery to parents who may be facing the same news?

Describe what it is like to live with the knowledge of your child being a carrier.

- What has it been like for you since finding out?
- Do thoughts related to your child being a carrier enter your mind much? Is there a particular aspect of it that you usually dwell on?

Tell me something about how your partner feels about your child being a carrier.

- Is it something you have discussed together much?

What is it like to know that [you have/your partner has] passed on this gene to your child?

- Tell me about the ways you may have come to terms with it.

Does your child know that they carry the CF gene?

⇒YES.

- What was it like to have to tell them yourself?
- How did you go about telling them/ broaching the subject with them?
- Did you anticipate your child reacting in any particular way? Compare this to how they actually did react.

⇒NO.

- Do you think you will be the one to tell them about it?
- Have you got any ideas about a particular time or situation when it will be best to bring up the subject?
- What do you think it will be like telling them?

Imagine that you're an expert in dealing with this whole experience of finding out your child is a carrier, living with that knowledge and then dealing with how and when you are going to tell them, what would you say to someone who had no idea about what it is like?

Is there any way your child's life would have been different so far if they did not carry the CF gene?

How do you think your child's future would be different if they did not carry the CF gene?
etc.

ADDITIONAL INTERVIEW QUESTIONS (OPTIONAL)

How did your family/friends/doctors/nurses make a difference?

Describe how you feel about the word 'carrier' itself? What's the difference between being a 'carrier' and being 'normal'?

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