# Part Three BENEFITS AND HARMS IN GENETIC TESTING OF MINORS


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# 1 PREAMBLE

Having looked at the issues raised by genetic testing of minors, and the professional guidelines, attitudes and practice in respect of such testing, it is apparent that much hangs on the perceived benefits and harms.

This part of the report is concerned with analysing the purported benefits and harms of carrier, pre-symptomatic and susceptibility testing for both childhood and adult-onset conditions, and the available evidence on the subject.

## 1.1 Terminology: Benefits and harms

This section focuses on the purported benefits and harms of genetic testing of minors. We use the terms 'benefits' and 'harms' rather than other dualisms, such as 'advantages' and 'disadvantages', for a number of reasons. First, 'benefit' easily accords with the bioethical principle of beneficence (the health professional's duty to do or produce good);[1] and, likewise, harm accords with the principle of non-maleficence, or *primum non nocere* (first do no harm).[2] They are thus appropriate lenses through which to consider the effects of genetic testing. Secondly, our legislation permitting minors of or over the age of sixteen years to make their own medical decisions explicitly uses the term 'benefit' to describe the kinds of procedures minors may consent or refuse to consent to (s 36(1), Care of Children Act 2004). It is thus essential that the effects of genetic testing are considered in terms of benefits and harms so that they can be analysed against our legislative framework for minors' decision-making. And, finally, benefits and harms appear to be the dominant construction through which to examine genetic testing of minors given that reference to benefits, and more usually harms, are rife in the medical genetics literature, and in the professional position statements and guidelines discussed earlier.

'Benefit' has a wide meaning. It covers a broad spectrum of advantages that may accrue to humans, including medical, physical, psychosocial, emotional and spiritual benefits.

We define a beneficial medical intervention (which may give rise to a 'medical benefit') as a procedure or treatment designed to cure a disorder; suppress the symptoms of a disorder; or ameliorate the symptoms of a disorder. Other types of interventions available in the health-care setting, such as counselling or helping blind patients learn how to read Braille or walk with a stick, are psychosocial benefits, aimed at helping a patient cope or adjust to a disorder, and not strictly 'medical benefits.'

There is great debate as to whether genetic testing of minors is beneficial or harmful, largely because of the limited empirical evidence on the effects of predictive testing

in minors in particular. Most of the major professional guidelines and position statements on genetic testing of minors take a cautionary stance, advocating abstention from the testing of minors generally (except where a medical benefit may accrue), in accordance with the perceived best interests of the minor.[3] But is genetic testing of minors as harmful as is claimed?

There is scant evidence of either psychosocial benefits or harms resulting from predictive testing of children. Thus, while there is little evidence that 'pre-emptive knowledge of future ill-health'[4] is a good, there is also limited evidence of the alleged harms it causes. There is more evidence relating to the effects of carrier testing in children, as these tests have been possible for much longer.

### 1.2 Going beyond the Huntington disease (HD) paradigm

Many of the earlier guidelines and commentaries on predictive genetic testing generally, and genetic testing of minors specifically, used testing for the Huntington disease (HD) mutation as an example or case study of potential benefits and harms.[5] However, the HD mutation is unusual among late-onset disorders in respect of its seriousness and complete penetrance:

> *… HD is a distinctive condition, terrible in its manifestations, progression and fatal outcome. Therefore, it is inaccurate to generalize from testing for genes for this disease to testing for all other adult-onset conditions.*[6]

Hogben and Boddington challenge the frequent use of HD as the exemplar of a late-onset disorder. They note that choosing HD as the standard in predictive testing discourse 'inevitably acts to construct the information generated by predictive testing (in general) as highly undesirable'.[7] Their special interest is the distinction drawn between carrier testing and predictive testing, and they criticise the role of the HD example in underestimating the effects of carrier testing 'as "less serious" through implied contrast'.[8]

This is one of the reasons why we consider and differentiate between the purported benefits and harms raised by the various kinds of genetic tests. Each condition for which testing is available, whether early or late-onset, or involving symptomatic, pre-symptomatic, susceptibility or carrier testing, has different features to some extent and thus the implications of testing will differ accordingly.

### 1.3 Minors who cannot give informed consent and competent minors

We deal separately with the benefits and harms arising from genetic testing of minors who request testing for themselves from the benefits and harms that may arise for those minors tested as a result of parental requests and consent. Different legal issues are raised for minors who seek testing themselves and those who are tested on the basis of parental consent – which is why the legal frameworks for medical decision-making in respect of each group of minors are also discussed separately in the following sections.

Additionally, one of the most frequently cited potential harms of genetic testing in childhood is that it infringes a minor's autonomy to make a decision about testing. Opponents of genetic testing of minors argue that predictive and carrier testing of minors infringes their autonomy and the right not to be tested and not to know their genetic risk status as adults. Thus, many of the arguments against predictive or carrier testing of minors disappear or, at the very least, are mitigated in the context of competent minors seeking testing for themselves.

Very little evidence is available of the actual benefits and harms that arise from genetic testing of minors. We discuss the benefits and harms of genetic testing of competent minors first largely because more evidence exists, thanks to Duncan's research,[9] of the outcomes of testing this group (compared to outcomes for those too young to give informed consent).

As Duncan states:

> *Of the five empirical studies that have been conducted in order to answer questions about the impact of such testing, not one has reported the opinions of young people. What do young people who have undergone predictive genetic testing think about the experience? … Do they feel harmed by the knowledge of their genetic status or are they glad to know? Do they simply feel ambivalent? We don't know. The vast majority of potential harms that are purported to be associated with predictive genetic testing in young people are psychosocial harms, not physical harms. These are therefore at least partly subjective. In order to assess these outcomes accurately, we must consult young people who have experienced predictive genetic testing themselves. They are the only ones capable of providing us with real insights into the effects that such testing has upon young people.*[10]

## 2 BENEFITS AND HARMS OF GENETIC TESTING IN COMPETENT MINORS

*A clash in perception is taking place. Some perceive predictive genetic testing in young people to be too potentially harmful to allow. Others perceive it to be an opportunity for benefit, even an opportunity for the prevention of harm.*[11]

### 2.1 Introduction

There is some evidence that minors occasionally request genetic testing.[12] Some seek it on their own behalf, while others do so in conjunction with their parents or guardians. However, there is considerable disagreement as to whether minors should be tested at all, regardless of their competence. The issue therefore is the harmfulness of genetic testing. This section will consider the potential benefits and harms of genetic testing of competent minors to determine the validity of the perception of harm.

Arguments opposing genetic testing in competent minors are generally premised on one or both of the following lines: the consequences of genetic testing may be too harmful to allow minors the ability to give legally effective consent to testing (even if they are competent to consent to other medical procedures); and genetic information and the implications of genetic testing are such that minors cannot competently comprehend them and give effective consent to testing. According to this view genetic testing could not be undertaken until the minor attained the age of eighteen.[13] In this section we consider whether there is merit in the view that minors should not be granted access to genetic testing, even upon a competent request, by examining the harms and benefits of genetic testing. The limited evidence available regarding harms does not support the view that the consequences of testing would be too harmful for competent minors who request testing. The evidence there is suggests that not only are there benefits in testing competent minors, but also harm may result from not testing competent minors who for good reason request a genetic test.

### 2.2 Benefits and harms for competent minors specifically

Much of the discourse and professional guidance on genetic testing and minors relates to minors too young to seek or give valid consent to genetic testing. Thus most of the debate focuses on parental rights with regard to their children's medical treatment, the minor's future autonomy and the best interests of the minor, including concerns about the potential misuse (whether intentional or otherwise) of genetic information. Many of the arguments against predictive testing of minors disappear or, at the very least, are mitigated in the context of competent minors who themselves are seeking testing.

Opponents of genetic testing of minors argue that predictive and carrier testing of minors infringes their autonomy and their right to not be tested and not to know their genetic risk status as an adult. Protecting a minor's 'future autonomy' to make a decision about whether or not to undergo genetic testing is also connected to protection of confidentiality. A minor who undergoes a genetic test in response to a parental request will generally not be granted the same confidentiality as an adult, given that the results will be disclosed to parents. These are among the most forceful arguments against predictive testing of minors, and are discussed in greater detail later.

Arguments opposing testing in competent minors are somewhat self-defeating to the extent that they rely on protecting a minor's autonomy and 'future autonomy'. Denying an autonomous competent minor's request for a genetic test in order to safeguard autonomy is a *non sequitur*.

> *The sincerity of the geneticist in seeking to protect the child's right to autonomy is not in question. The issue is whether the decision not to test effectively abrogates what it seeks to protect, the child's personal rights and dignity subordinated to, if not replaced by, the objectives, values, and rationality of the geneticist.*[14]

Additionally, where a minor is deemed competent to make a decision about genetic testing, the minor will also be entitled to confidentiality, as an autonomous individual.

The benefit or harm of a genetic test is likely to be primarily subjective: does the competent minor who is seeking a genetic test consider that the test will be beneficial? The following factors, among others, may be relevant to the minor: the kind of genetic test in question; the kind of disorder being tested for; and the reasons why the test is being sought. For these reasons the analysis of benefits and harms is divided under headings relating to the different kinds of genetic tests available (as outlined in Tables 1 and 2).

The focus is on the potential benefits and harms of genetic testing for minors who are competent and who request testing for themselves.

### 2.3 Purported benefits and harms of different types of genetic testing

> *The range of social differences between young people and adults means that it is likely predictive genetic tests will impact differently upon young people from the way in which they impact upon adults. However, it remains unclear if these differences will result in a greater potential for harm or greater potential for benefit when young people undergo such testing.*[15]

### 2.3.1 Symptomatic testing

Symptomatic genetic testing for a disorder for which there is treatment available is generally considered to be beneficial: once the diagnosis is established, the appropriate treatment programme can begin. What about symptomatic genetic testing for a disorder for which there are no beneficial medical interventions available? Symptomatic testing is generally regarded as beneficial regardless of whether or not the disorder tested for can be medically managed. Benefits include: the cessation of appointments, procedures and tests (some of which might be invasive or harmful) in order to ascertain what is causing the symptoms; and the reassurance of knowing what is wrong and what to expect in terms of progression of the disorder, and therapeutic or other support.

In cases such as these, where a specific genetic mutation is suspected, the health professional involved would almost certainly urge genetic testing because of the belief that the benefits of identifying the etiology (cause or origin) of the symptoms far outweigh any harms that might arise from having a clear diagnosis.[16]

We therefore endorse Clarke and Flinter when they state that:

> *There may be ethical implications arising from the diagnosis of certain specific conditions, or from attaching a potentially stigmatising diagnostic label to a child … These problems, however, are of a general nature, are not specific to molecular genetic testing, and will not be addressed further …*[17]

The purpose and principle of symptomatic genetic testing is routine: the health professional is searching for the cause of symptoms (specifically seeking to establish whether they might have a genetic basis), as they would for any type of disorder via other methods of clinical examination or testing. The ethical implications of diagnosing certain conditions or attaching a potentially stigmatising diagnostic label to a child could arise equally in diagnosis of a child with HIV/Aids or other conditions. Other methods of symptomatic examination or testing can also have implications for family members, e.g. any form of examination or testing which indicates a heritable genetic mutation, or testing for any communicable virus such as HIV/Aids or hepatitis.

### 2.3.2 Predictive testing for late-onset disorders[18]

#### 2.3.2.1 Pre-symptomatic testing for late-onset disorder for which there are beneficial medical interventions

If pre-symptomatic surveillance or interventions have a potential medical advantage then there are clear benefits to predictive testing. The genetic information allows the person and their health professional to implement interventions at the earliest possible opportunity.

An example of an autosomal dominantly inherited genetic condition for which pre-symptomatic testing and pre-symptomatic beneficial medical interventions are available is familial adenomatous polyposis (FAP).[19] A person with the inherited mutation for FAP develops many polyps in their bowel, generally by their late teens. Without treatment some of these polyps will develop into cancer. Bowel cancer due to FAP often occurs in the third, fourth or fifth decade of life, but sometimes even earlier.[20] When a person is identified as having the mutation, regular surveillance via colonoscopy can detect the appearance and growth of polyps, potentially allowing them to be removed before cancer develops; or detect the early signs of cancer, allowing treatment to begin at the earliest possible opportunity (surgery to remove the cancer or bowel, radiotherapy or chemotherapy). More than 90 per cent of bowel cancers can be cured if picked up at the earliest stage.[21] There is thus potentially great clinical benefit in pre-symptomatic testing for the genetic mutation responsible for FAP, and other disorders for which there are pre-symptomatic beneficial medical interventions available.

If a disorder cannot be prevented or delayed and there are no medical interventions available before symptoms appear then there may be no clinical benefit to testing being undertaken pre-symptomatically (even if there are interventions available once symptoms appear). That is, testing can be delayed until a person becomes symptomatic.

And yet there may be other non-medical benefits to pre-symptomatic testing for a genetic disorder for which there is treatment available (either before or after a person becomes symptomatic). Many of these may accrue regardless of whether the test results are positive for the genetic mutation, or mutation negative.

However, pre-symptomatic testing for a disorder for which there are medical benefits available (whether pre- or post-symptomatically) also has the potential to cause harm.

See Tables 1 and 2 for a list of the purported non-medical benefits and harms in relation to pre-symptomatic genetic testing and various test results.

***Table 1:*** *Purported non-medical benefits of pre-symptomatic genetic testing of minors for conditions for which beneficial medical interventions are possible*

| Potential benefits regardless of test result | Potential benefits more specifically associated with mutation-negative result[37] | Potential benefits more specifically associated with mutation-positive result |
|---|---|---|
| – Enhanced autonomy: minors develop their competence, confidence and autonomy by making increasingly important decisions about their lives for themselves. A minor needs the opportunity to practice and refine decision-making skills: testing helps to increase the sense of the self as an active participant in life, rather than a powerless victim of adults and genes.[22] Granting choice and control has a positive value for adolescents and respecting their choices has positive consequences for their self-esteem and psychological health.[23] The genetic testing process can empower minors.[24]<br>– The self-knowledge gained from the test results can also 'promote more autonomous decision making about one's life'.[25]<br>– Confronting pre-symptomatic testing can result in a minor moving through the 'rebellious teenage' stage more quickly.[26]<br>– Testing relieves the uncertainty and anxiety of not knowing whether one carries a genetic mutation that will result in a late-onset disorder.[27] It may remove a mental block, and eradicates one of the unpredictabilities associated with the condition for which one is being tested.[28] | – Knowing that the genetic condition will not develop.<br>– Being able to stop worrying.<br>– Relief from anxiety about possible early signs of disorder.[38]<br>– Feeling freer, able to plan and excited about the future.[39]<br>– Positive psychological effects include enjoying life more; feeling more outgoing; feeling more normal; feeling happier; increased self-esteem; and feeling as though a new life has been given.<br>– Feeling that children are a possibility and not having to worry about them developing the condition.<br>– Knowing that partners will not be burdened by the genetic condition in the future.<br>– Allowing parents to stop worrying and seeing parents happy.<br>– Feeling more able to help gene-positive parents. | – Establishing new relation-ships with people in similar situations.[41]<br>– Allows time for research and adjustment.[42] |

– Testing involves a direct encounter with a trained professional who can transmit accurate information directly and supplement complete reliance on parental transmission of information.[29]

– Testing minors at their request allows them to psychologically adjust to the information at an age-appropriate time. It has been argued that 'being informed of a serious illness at an early age may facilitate adjustment and coping'.[30]

– Testing gives the minor the opportunity and time to plan for the future in view of the genetic information (including career and financial planning, and reproductive and end-of-life decisions).[31]

– Testing process can improve relationships with family members, including parents and siblings, as a result of opening communication; having a reason to spend more time with family members; realising how strong parental support is; and helping siblings through the same process. [32]

– Testing process can also improve and strengthen friendships.[33]

– More accurate genetic counselling becomes possible.[34]

– Testing provides opportunity to deal with many issues in the genetic counselling sessions.[35]

– Testing whilst a minor can mean that the person tested is young enough for the results not to have great impact.[36]

– Knowing that the condition stops now in the family.

– Cessation of being treated as if one did have the condition.[40]

***Table 2:*** *Purported non-medical harms of pre-symptomatic genetic testing of minors for conditions for which beneficial medical interventions are possible*

| Potential harms regardless of test result | Potential harms more specifically associated with mutation-negative result[37] | Potential harms more specifically associated with mutation-positive result |
|---|---|---|
| – Testing process and test results can mean taking time off school and can interfere with school work.<br>– Anxiety and stress whilst waiting for test results.<br>– Counselling prior to testing and disclosure of test results dragging on.<br>– Regrets about being tested.<br>– Negative psychological effects after receiving test result such as feeling flat or lost and not knowing how to proceed.<br>– Wanting more contact with the counsellor than provided after receipt of test result.<br>– The information may be misunderstood which can lead to misconceptions about the future.<br>– Being too young to understand what the test entailed.<br>– The possibility of mistaken paternity being identified.<br>– Testing during adolescence might interfere with natural separation process that needs to occur between parents and children, because of a bond between | – Believing that one's own test result implies a specific result in a sibling.<br>– Feeling concerned about affected siblings.<br>– Feeling concerned about having to look after an affected sibling or parent later in life.<br>– Feeling guilty about not being affected when siblings or parents are affected. | – Knowing that the genetic condition could be fatal[54]<br>– Negative psychological effects, such as worry about the onset of the genetic condition; feeling shocked; feeling angry; feeling depressed; feelings of despair; thinking 'why me?';[55] low self-esteem; or serious psychological maladjustment.[56 57]<br>– The adolescent process of establishing a personal identity might be made particularly difficult by a mutation-positive test result.[58]<br>– Watching parents become upset.[59]<br>– Watching a symptomatic parent as an affected individual.<br>– The test result resurfacing when other things go badly in life.<br>– Concern about not being able to perform at work, or about being fired.<br>– The minor and/or family might develop a perception that the minor is already ill and vulnerable or a victim[60] (dietary, environmental or other 'health' interventions might be implemented in an effort to stave off the disorder; or fewer resources might be expended on the minor). |

| | | |
|---|---|---|
| an affected minor and affected parent, or because of guilt over not being affected when one's parent is.<br>– Feeling distanced from parents who do not share the same genetic status.<br>– Drifting apart from parents.<br>– Feeling angry about manner in which and to whom results were disclosed.<br>– Anxiety about others learning test result, and their possible reactions and potential for gossip.<br>– Being treated differently and feeling let down by unsupportive friends.<br>– Desire for time alone affecting friendships.<br>– Having to provide explanations about genetics to friends. | | – Minor might be discriminated against or scapegoated within the family because parents are reminded of their own unacceptable traits.[61]<br>– Affected minor may be forced into the realisation that her parents are not 'all-powerful' and weare unable to prevent the genetic condition. This could be psychologically damaging if occurring too early in life.[62]<br>– Minor may not want to identify with sick family members.[63]<br>– Disclosure of test results to others, including family members, resulting in loss of privacy.[64]<br>– Stigmatisation in the family and community, with reduced opportunities for education, marriage and reproduction.[65]<br>– Minor might find it difficult to integrate with peers because of feeling different, and not desirable for dating or marriage.[66]<br>– Parents may not be able to help with the impact of the test result because of their own issues, or guilt over genetic inheritance.[67]<br>– Social discrimination regarding, for example, employment and access to insurance.[68] |

There are thus a variety of purported benefits and harms related to pre-symptomatic testing for a disorder for which beneficial medical interventions are available.

The magnitude of some of the potential benefits and harms listed in Tables 1 and 2 will be affected by the type of medical benefit available for the particular disorder for which the minor has been tested. The potential psychological harm involved in discovering one will manifest a disorder later in life may be minimised if the disorder is curable, as opposed to a debilitating, degenerative disorder for which the only beneficial medical intervention available is treatment to ameliorate the symptoms.

#### *2.3.2.3 Pre-symptomatic testing for late-onset disorder for which there are no beneficial medical interventions*

Is a pre-symptomatic test for a late-onset disorder, for which there are no beneficial medical interventions available, beneficial or harmful to a competent minor requesting such a test? Where there are no pre-symptomatic, beneficial medical interventions available (no prophylaxis, no cure and no treatment which will suppress or ameliorate the symptoms) then there is no medical benefit to pre-symptomatic testing.[69]

HD is an example of an autosomal, dominantly inherited, late-onset disorder for which there are no beneficial medical interventions available. HD is a neurodegenerative condition with an age of onset generally between thirty and fifty. Early symptoms include involuntary movements in the body and limbs, personality changes including irritation, poor insight, depression, withdrawal, euphoria and difficulty with organisation. Eventually, speech becomes slurred, swallowing difficult and walking unsteady. HD sufferers often succumb to a respiratory illness or complications from falls.[70]

Despite the fact that there are no clinical benefits to testing, the same potential non-medical benefits may accrue as those listed in respect of pre-symptomatic testing for a disorder for which there are medical interventions available (such as enhanced autonomy; reduced uncertainty; direct encounter with a trained professional; time to adjust; opportunity and time to plan for the future around the genetic information; and improved and strengthened family relationships and friendships).

However, it may be argued that there is greater potential for harm in respect of testing for a disorder for which there are no possible medical interventions compared with testing for a late-onset disorder for which medical interventions are available. The same harms as listed may be consequent on such testing, but with a greater likelihood of more psychological harm: almost all of the purported harms listed might be exacerbated by the knowledge that no beneficial medical interventions are available. Knowing one will develop an entirely untreatable disorder later in life could result in extreme despair or depression, and 'cause worse problems than continued uncertainty'.[71]

Additionally, an early diagnosis could result in increased tests, monitoring and treatment regimens (at a cost) with no proven benefits.[72]

#### *2.3.2.3 Susceptibility testing for late-onset disorder for which there are beneficial medical interventions*

Is a susceptibly test for a late-onset disorder that can be treated beneficial? There may be pre-symptomatic clinical benefits to such testing. For example, testing for familial hypertrophic cardiomyopathy (associated with increased risk of sudden death) allows drug therapy to prevent arrhythmias;[73] and for a number of conditions (e.g. familial hyperlipidemia) preventative advice can be given about lifestyle changes which have therapeutic advantages.[74] Predictive testing for the genetic mutation that predisposes people to haemochromatosis (chronic disorder involving the excessive absorption and inappropriate storage of iron)[75] can yield beneficial clinical results: organ damage can be prevented by beginning venesection (the removal of blood) as soon as possible. The earlier that venesection begins, the better the prognosis. People who are diagnosed with haemochromatosis and who begin venesection prior to developing liver damage can enjoy a normal life expectancy.[76]

Predictive testing for a BRCA 1 or BRCA 2 mutation that predisposes a woman to breast and ovarian cancer might also have useful clinical benefits including increased surveillance by the woman and her health professional, or even prophylactic surgery, where such is deemed appropriate. The earlier a cancer is found the more successful the treatment is likely to be.[77]

Many of the disorders for which susceptibility testing can be undertaken are multifactorial. That is, there are a number of factors that influence whether or not the person will develop the disorder: a faulty gene is not the only cause of the disorder. There may be more prophylactic measures available than for genetic mutations with 100 per cent penetrance, e.g. dietary, environmental or other lifestyle changes or surgery to remove potentially affected areas (e.g. breasts, ovaries or bowel) to minimise the risk of developing the disorder. Surveillance aimed at early detection may also have great potential to minimise the harm caused by the disorder. However, a great deal remains unknown about the other factors that trigger the onset of the types of disorders discussed.

In terms of non-medical benefits, similar issues arise as for *pre-symptomatic* testing for late-onset disorders for which there are available medical interventions. Many of the same benefits may accrue.

> *For a rational person wishing to pursue rational life plans, knowing whether she is carrying a BRCA 1 mutation can be a major benefit because in our society important decisions about career and life orientation have to be made between*

*the ages of 14 and 17 years. A minor may also wish to decide about becoming pregnant. Knowing that she is a BRCA 1 mutation carrier, she might decide against a long university education to have children early and pursue a university career after having had bilateral mastectomy and oophorectomy.*[78]

However, some of the potential benefits gained from pre-symptomatic testing (i.e. for mutations with 100 per cent penetrance) are reduced or do not apply in respect of susceptibility testing. Susceptibility testing cannot remove the uncertainty to the same extent, unless the result is mutation negative. Even if a person tests positive for the mutation in question then it is still not known whether or not the disorder will develop – only that there is increased risk. This lack of certainty also reduces the potential degree of benefit in being able to plan one's future around the genetic information revealed.

Similar harms might be attendant upon susceptibility testing for treatable disorders as those that arise in the context of pre-symptomatic testing. Additional harms may also arise in the context of susceptibility testing. The fact that the person may never develop the disorder adds a unique layer to the potential harms: many of the harms listed may be suffered unnecessarily. That is, the person's psychological health and happiness, chances of obtaining life or health insurance and employment prospects may all be affected because of a susceptibility to a disorder that may never develop.

*Unpatients may develop severe anxiety and believe that they are under a death sentence. They may visit doctors frequently, seeking monitoring and reassurance. In extreme cases, they may contemplate suicide. They may be ostracized by friends and family and be discriminated against in insurance and employment.*[79]

However, the purported harms involved in susceptibility testing for a treatable late-onset disorder may be mitigated by the knowledge that one may never develop the relevant disorder: a positive test result for the genetic variation is not a definite sentence regarding the disorder (as it is in the case of pre-symptomatic testing).

Again, the magnitude of the potential benefits and harms will be affected by the type of medical benefit available for the particular disorder: amelioration or suppression of symptoms, or cure. The degree of susceptibility to the disorder will also be relevant: there may be more harm attendant upon knowledge of a 90 per cent chance of developing the disorder, for example, than a 20 per cent chance.

Penetrance is often age related. For example, it is estimated that the cumulative risk of breast cancer for women with BRCA 1 mutations is around 3.2 per cent by age thirty, 16 per cent by age forty, 59 per cent by age fifty and 82–85 per cent (approximately) by age seventy.[80]

#### *2.3.2.4 Susceptibility testing for late-onset disorder for which there are no beneficial medical interventions*

Is a susceptibility test for an untreatable late-onset disorder beneficial? Where there are no pre-symptomatic, beneficial medical interventions available (no prophylaxis, no cure and no treatment which will suppress or ameliorate the symptoms) then there is no medical benefit to susceptibility testing.

Alzheimer's disease is an example of a multifactorial late-onset disorder (which is thought to have a heritable genetic basis) for which there are only very limited beneficial medical interventions available (medications to slow the decline). Alzheimer's disease involves 'gradual memory loss, decline in ability to perform routine tasks, disorientation in time and space, impairment of judgment, personality change, difficulty in learning and loss of language and communication skills'.[81] The age of onset and rate of disease progression varies, and the life span from onset can be anywhere from three to twenty years. The disease eventually leaves sufferers unable to care for themselves.[82]

Some of the same potential non-medical benefits may accrue as for pre-symptomatic testing for the other kinds of disorders: the positive psychological effects of a mutation-negative result; testing pursuant to the minor's request enhancing autonomy; and testing enabling psychological adjustment.

Fewer non-medical benefits may apply in respect of susceptibility testing for an untreatable late-onset disorder. Unless the result is mutation negative, the anxiety involved in 'not knowing' may not be mitigated and the potential benefit in being able to plan better for the future is reduced.

Similar harms might be attendant upon susceptibility testing for an untreatable late-onset disorder as for pre-symptomatic testing for an untreatable late-onset disorder. There may be greater potential for psychological harm in respect of testing for a disorder for which there are no medical benefits. Knowing one is at a significantly increased risk for an untreatable late-onset disorder could result in extreme despair or depression.

Again there is the additional mischief that these more severe harms may be suffered unnecessarily, as the person may never develop the disorder.

Conversely, the harms involved in susceptibility testing for an untreatable late-onset disorder may be somewhat mitigated by the knowledge that one may never develop the relevant disorder: again, a positive test result for the genetic variation is not a definite sentence (as it is in the case of pre-symptomatic testing).

The magnitude of the potential benefits and harms may be affected by the degree of susceptibility to the disorder. There may be more harm in knowing that one has a 90 per cent chance of developing the disorder as opposed to a 20 per cent chance.

### *2.3.3 Carrier testing*

It is important to distinguish carrier screening from carrier testing, at this juncture. This section of the report focuses on testing of 'at-risk' individuals, and not screening of large groups or populations. Carrier screening usually refers to testing programmes involving large groups of people, who may or may not be individually 'at risk' for the disorder tested for,[83] and raises political, legal, cultural, social and economic issues that do not necessarily arise in respect of testing 'at-risk' individuals for carrier status.

For the individuals tested, there are generally no medical benefits to carrier testing.[84] However, the test results do allow the individual to make more informed reproductive choices.[85] Many of the purported benefits are also relevant to carrier testing (with a change in terminology and shift in focus from developing a disorder to knowing one's offspring may develop a disorder). The benefits relating to future planning, particularly around reproductive decisions, are of great significance in the carrier-testing context. The purported benefit of being tested at an age-appropriate time is also of particular weight. The competent minor requesting testing may well be considering embarking on a sexual relationship, and may require knowledge of carrier status in order for sexual behaviour to be as informed as possible.

Fanos argues that siblings of affected individuals may view their carrier status as a benefit in terms of being able to feel a part of the disorder community, like others in the family, and feeling closer to the sibling with the disorder. Additionally, carriers may feel less survivor guilt if they perceive that they are sharing the burden of the condition.[86] Being a carrier means that the 'unaffected' individuals are also affected to a certain extent (usually primarily in respect of reproductive choices) by the disorder and thus may not feel as guilty as if they had entirely escaped the burden of the disorder from which their siblings suffer.

However, that is not to say that psychosocial harms do not exist in respect of carrier testing. The possibility of feeling, or being, stigmatised is real, and carrier testing may have a unique ability to evoke fear of intimacy and interpersonal relationships, relating as it does to sexual relations and reproductive choices.[87]

Additionally, it has been suggested that carrier testing may lend itself to particular potential for misunderstanding the implications of the test results, being a medical test 'not linked to discovery of illness'.[88]

*Given symptoms, an individual will seek a diagnostic label, and given a label, he or she will seek symptoms.*[89]

Some consider carrier testing to be less serious, or less harmful, than predictive testing of minors,[90] because the individual tested will not develop the disorder, and is 'only' at risk of passing on the disorder.

Some of the purported harms are not applicable or are arguably less threatening in respect of carrier testing. For example, knowing that the genetic condition, for which one has tested positive, could be fatal is not strictly applicable to unaffected carriers (although it may be to their potential offspring); watching a symptomatic parent as an unaffected carrier may not be as psychologically harmful as watching a symptomatic parent as an affected individual given that one will not develop the disorder oneself; access to and concerns regarding education, employment and insurance are not as salient; and it is arguably less likely, if everybody is adequately informed, that a carrier will be treated as vulnerable, or as a victim, than will be the case for a pre-symptomatic affected individual.

Hogben and Boddington take issue with the distinction frequently drawn between testing minors for carrier status, and testing them for late-onset disorders.[91] They argue that the distinction rests on an assumption about the status of reproduction in people's lives, and on an ethical stance that prioritises the self over others.[92] They criticise the implicit assumptions that reproductive issues are 'of relatively minor import' compared with issues related to one's own health status; and that issues that concern others (such as the 'next generation') are relatively minor compared with issues that concern oneself. They argue that carrier status may be of profound importance for certain people, and for certain disorders.

*Some may rank the threats to parenthood posed by carrier status as higher than the threat to their own health status. … It is important to be reflective about how much the view that carrier status is of relatively minor importance is shaped by the assumption that carrier status problems can be 'solved' with the use of genetic testing and technology.*[93]

Hogben and Boddington highlight the fact that the focus on the implications of carrier testing for reproductive decisions means that 'the potential implications for the reproductive self, which result from predictive testing, are routinely overlooked'.[94]

## 2.4 Purported harms of not testing on a competent minor's request

Clinical or physical harms can arise partly as result of not having a genetic test. For example, a person affected by the FAP mutation will almost certainly develop cancer if the polyps in the bowel are not detected early enough. In cases where there is a clear medical benefit to predictive testing, and conversely likely harm consequent upon not testing, one would expect that testing would go ahead. That issue is relatively non-controversial. But there may also be harms in not performing tests where the medical benefit is not so clear-cut.

Professional guidelines, position statements and opponents of genetic testing do not often refer to the potential (non-medical) harms that may arise from refusing to provide a competent minor with a predictive genetic test upon request.[95]

> *What are noticeably absent, however, from such arguments are the child's … reasons for wishing to undertake the procedure. 'To provide benefit and to prevent harm' apparently has been assessed primarily from the geneticist's perspective. What of the harm which may result from not administering the procedure? What of the depression and anguish a child could experience due to uncertainty as to risk status?*[96]

### *2.4.1 Potential non-medical harms of not testing – All kinds of genetic testing*

Many of the harms that may potentially arise from not testing a competent minor who is requesting a genetic test are the converse of the potential benefits. For example, refusing to test may:

- Restrict development of and diminish confidence, decision-making ability, competence and autonomy;
- Limit the minor's self-knowledge (which would have been enhanced by information given via the test result) and thereby further diminish autonomy and decision-making capabilities;
- Prolong the uncertainty and anxiety of not knowing whether one carries a genetic mutation that will result in a late-onset disorder or that one could pass on to one's offspring;
- Limit the time available to adjust psychologically to the information at an age-appropriate time;
- Deny minors the opportunity and time to plan their futures around the genetic information (given that many minors make career and other life plans in adolescence);
- Prolong family and friendship tensions;

- Limit the opportunity for reflection, communication and information gathering that is encouraged in genetic counselling;
- Delay the psychological benefits consequent upon finding out one does not have the genetic mutation tested for;
- Delay reproductive decisions; and
- Prolong concerns about how partners, potential partners, parents, siblings, friends, family members and wider groups might react to the test result.

Other harms may arise from not testing, which amount to more than simply the denial of the potential benefits referred to earlier.

For example, refusing to test may:

- Frustrate the minor seeking testing and result in feelings of unworthiness;
- Result in or prolong risk behaviours in minors who know that they are at risk for a heritable genetic condition;
- Prolong the period in which minors assume and act as though they have the genetic mutation. Living as if one has a condition can bring with it almost all of the harms associated with actually having the condition.[97]

> *… the decision to postpone testing can never be taken lightly. The stress of undergoing testing, receiving a result, and adjusting to the new risk status must be weighed against the stress and uncertainty of living at risk for HD, the blow to the candidate's self-respect by being denied testing and the possible sense of humiliation and helplessness by having one's autonomy undermined.*[98]

As with the purported harms and benefits of genetic testing, there is a lack of empirical evidence regarding the purported harms of not testing, especially in minors. The existing evidence of harms and benefits and the relevance of a lack of such evidence are considered in the next section in the context of each type of genetic test.

## 2.5 Evidence of benefits and harms, or lack thereof

### *2.5.1 Predictive testing*

There is a little evidence-based information about the psychological and social effects of predictive genetic testing in children, including competent minors.[99] Much of the discussion is based on speculation: some of the purported benefits and harms are imported from research involving predictive genetic testing of adults, and some are analogised from other medical contexts.[100]

There have been very few studies into the psychological and social effects of predictive testing in minors. Of the few that exist, some refer to particularly small case studies involving one or two very young children, and relate mostly to parental responses to testing.[101]

Johnson studied the emotional and cognitive effects of testing for a diabetes predictor on fifteen children and twenty-one adults. She found that children, like adults, exhibited clinically significant anxiety when first told they were at risk, but returned to normal levels within two or three months.[102]

A Canadian team of researchers studied the behavioural effects of testing minors aged four to seventeen years for hyperlipidemia (elevated or abnormal levels of lipids or lipoproteins in the blood).[103] While no differences were found in anxiety, depression, social competence or behavioural adjustments between the thirty-four minors who tested positive and the twelve who tested negative, mothers reported more behavioural problems in the positive children.[104] However, as Michie and Marteau point out, it is unclear the extent to which reported behaviour reflected actual problem behaviour, or increased sensitivity on the part of parents.[105]

There are larger studies relating to predictive testing of young people for the FAP mutation.

Codori et al. studied the short-term psychological effects of genetic testing for FAP by surveying forty-one children (aged six to sixteen years) and their parents, before testing and three months after testing.[106]

Minors' mean scores for depression, anxiety and behavioural problems were in the normal subclinical range both before and after testing, 'suggesting that children tolerated genetic testing for FAP without clinically significant increases in psychological distress or deterioration in adaptive behaviour'.[107] Indeed, all minors showed a significant decrease in behavioural problems following testing.[108]

Minors with affected mothers had subclinical yet significant increases in anxiety, regardless of their own test result. They also had subclinical but significant increases in depression if their mothers were affected and they themselves received a gene-positive result.[109]

Michie, Bobrow and Marteau addressed two questions in their study into the emotional impact of predictive genetic testing for FAP in children and adults: (1) Do children or adults receiving positive test results experience clinically significant levels of anxiety or depression? (2) Do children receiving positive test results experience higher levels of anxiety or depression than adults receiving positive results?[110]

They measured the anxiety and distress levels of sixty minors (aged ten to sixteen) who were tested for the FAP mutation, and compared them with the same measures in 148 adults (aged seventeen to sixty-seven) who were also tested for the same mutation.

They found that the children (both those who received mutation-positive results and those who received mutation-negative results) had depression and anxiety levels and behavioural problems within the normal range. In contrast, 43 per cent of the adults who received mutation-positive results displayed clinically significant levels of anxiety.[111]

The authors argue that some possible explanations for the anxiety difference between children and adults are not supported by the data. Some might suggest that the children did not understand the meaning of the positive test result. However, children with positive results perceived a higher chance of getting polyposis, worried more about that chance and were more threatened by their test results than those who received negative results. Others might argue that the children did not perceive polyposis to be as serious as the adults did. However, there was no difference between children and adults as to how bad they considered polyposis to be, or how bad they thought it would be if they developed it.[112]

Another possibility was that children might express their anxiety behaviourally, rather than by self-report. However, parents' reports did not suggest that mutation-positive children had any more behavioural problems than average. The authors suggest that children may have less understanding of the social implications of the test result, such as obtaining a mortgage or insurance, and the reproductive implications. Alternatively, they may find the implications less threatening because they will arise in the future. However, the data did not support that contention either. Mutation-positive children did find their results more threatening than mutation-negative children; and Codori et al. found that, among children aged six to sixteen, age was not associated with anxiety, depression or behavioural problems.[113]

With regard to the course of anxiety in children over the first year after testing, children who received mutation-positive results did not show increased emotional distress after receiving their results, and their anxiety, depression and self-esteem scores were within the normal range and did not change over time. There were trends for children with positive test results to be more anxious and depressed than those who received negative results, and for a higher number of the former to be in the clinical range of anxiety. Minors who received a negative test result displayed decreased anxiety and increased self-esteem, and worried less about their chance of getting polyposis.[114]

The authors conclude that:

> *The results of this study suggest that in the short term at least, there are no adverse psychological consequences for children undergoing predictive testing for FAP when testing is offered as part of a clinical genetics service, which typically includes at least a pre-test and a results disclosure visit. Among adults, receipt of a positive test result is associated with clinically significant levels of anxiety.*[115]

In a further study Codori et al. assessed symptoms of depression, anxiety and behavioural problems in forty-eight minors between the ages of five and seventeen years (forty-one of whom had also participated in the study outlined earlier) tested for the FAP mutation, for up to four-and-a-half years after testing. Twenty-two of the minors were mutation positive, and twenty-two were mutation negative.[116]

Across all follow-up periods there were no clinically significant changes in the minors' psychological tests scores, regardless of test results. Behavioural problems decreased for all children.[117]

This study added to the factors analysed in the previous study by assessing the effect of a mutation-positive sibling on the children's psychological outcomes. There were subclinical, but significant, increases in depression symptoms amongst minors who tested positive and also had a mutation-positive sibling, and several mutation-negative children with mutation-positive siblings displayed clinical elevations in anxiety.[118]

Duncan contends that this '(minimal) body of empirical research … reports a largely beneficial group of experiences' as a result of predictive testing of minors for the FAP mutation, including decreases in anxiety and behavioural problems.[119]

However, Duncan criticises the narrow range of potential benefits and harms focused on in studies about predictive testing in minors (depression, anxiety and behavioural problems), and further contends that the majority of outcomes researched have been harms of testing, rather than benefits.

*Therefore, not a single empirical study has been conducted with the specific aim of measuring benefits of predictive genetic testing in young people. … If we seek only to document the harms associated with testing, harms are all that we will find.*[120]

Pre-symptomatic genetic testing for HD has been available for longer than predictive testing for any other heritable genetic disorder,[121] giving rise to studies on the longer-term implications of testing.[122]

Duncan reviews the literature on the effects of predictive testing of adults for HD, to gain an impression of the possible effects of pre-symptomatic testing of minors for untreatable late-onset disorders (as opposed to pre-symptomatic testing for FAP, which has medical benefits).[123]

Regarding evidence of predictors of how individuals will react after predictive HD testing: individuals' negative feelings prior to testing (e.g. avoidance of HD-related situations, and dissatisfaction) were often exacerbated post testing (e.g. avoidance behaviour, depression, suicidal thoughts); although feelings of anxiety were often assuaged after testing. 'Test result, age and gender have been shown to be bad predictors of reactions after testing.'[124]

Duncan concludes that fears that predictive testing for the HD mutation would have catastrophic effects have not materialised.

*… in the short-term … those receiving a gene-positive test result are likely to experience a range of unfavourable outcomes. However, over time these dissipate, leaving little difference in the psychological functioning of those receiving gene-positive test results and those receiving gene-negative test results. In fact … both … have demonstrated decreases in psychological distress.*[125]

However, Duncan does note the need for caution in applying the results of the studies: all of the people studied were persons who voluntarily underwent the predictive test. Those people might be more psychologically resilient than others who choose not to undergo testing, or those who dropped out of the studies (recent research having revealed that those who drop out of research studies generally have higher levels of psychological distress, evaluated prior to drop out). Additionally the most long-term research results published indicated that pessimism may begin to grow amongst those with mutation positive test results, as the onset of the HD approaches. However, Duncan argues that this pessimism may in fact be related to the early symptoms of HD, so further research was required.[126]

*Taken as a whole, the body of research that describes the impact of predictive genetic testing for FAP in young people and for HD in adults tells a positive story. … Extrapolating the findings from these two similar fields could therefore lead us to believe that the likely effects of testing in young people may also be largely*

*beneficial. We may also extrapolate these findings to assume that predicting a young person's reaction to a test result may prove difficult … Until empirical research is performed concerning the impacts of predictive genetic testing in young people for non-medical reasons, the clash of opinion that is currently playing out in the academic arena will remain at the impasse it has reached."*[127]

#### *2.5.1.1 Predictive genetic testing of young people for non-medical reasons*[128]

Duncan et al. surveyed 301 respondents from the United States, Canada, the United Kingdom, Australia and New Zealand who were professionally involved in predictive genetic testing. They were questioned as to their involvement in relation to predictive genetic testing of minors for untreatable conditions, including any follow-up pursued post testing, and outcomes recorded.

Thirty-six clinicians (12 per cent) had been involved in providing predictive genetic tests for untreatable conditions to young people. There were forty-nine cases of such testing, in relation to fourteen different conditions. Of the twenty-seven cases (55 per cent) of testing of mature young people (fourteen or older), the majority (eighteen) involved minors aged sixteen to seventeen.

The most common condition young people were tested for was HD (fourteen cases). Other conditions tested for included myotonic dystrophy (the most common adult form of muscular dystrophy),[129] breast cancer predisposition and spinocerebellar ataxia ('characterised by slowly progressive incoordination of gait and often associated with poor coordination of hands, speech, and eye movements').[130]

In four cases (15 per cent) the young person's parents requested the testing, and in ten cases (37 per cent) the young person requested testing. In the remaining thirteen cases (48 per cent) the request for testing came from both the young person and the parents. Results were disclosed to all of the mature young people, except for one intellectually disabled fourteen-year-old.

Two thirds (18/27) of the cases of mature young people who had been tested were followed up, in a variety of ways. Two adverse events were reported: one seventeen-year-old male found to be at increased risk for HD was initially depressed and rebellious, but eventually accepted the result; and one seventeen-year-old female who was free of the HD mutation felt 'worry and responsibility for affected mother and untested brothers'.[131]

There were also nine reported beneficial effects: six following decreased risk results ('enabled him to focus on school', 'negative result really helped to stabilise life'); and three following increased risk results (e.g. 'So far she is doing fine and seems to have integrated this information into her thoughts about her future in a healthy way' and 'Coped better than many twenty-year-olds').[132]

#### *2.5.1.2 Duncan – Holding your breath: Predictive genetic testing in young people*

Duncan interviewed ten young people who had undergone predictive testing for FAP (participation rate of 20.8 per cent) and eight young people who had undergone predictive testing for HD (participation rate of 38.1 per cent).[133]

Of the ten respondents interviewed regarding testing for FAP, four were male, six were female, half were mutation positive, and half mutation negative. At the time of testing their ages ranged from ten to seventeen, and at the time of interviewing their ages ranged from fourteen to twenty-five.

Of the eight respondents interviewed regarding testing for HD, four were male, four were female, two were positive for the HD mutation and six were negative. At the time of testing their ages ranged from seventeen to twenty-five, and at the time of interviewing their ages ranged from twenty to twenty-six.

Duncan is well aware that 'The fundamental difference between these two types of predictive genetic tests is the existence (or lack) of medical benefit as a consequence of the test'."

> *When young people undergo predictive genetic testing for FAP they do so because, if found to be gene-positive, they are able to engage in preventative measures including screening and surgery to avoid the development of colon cancer. … Alternatively, when young people undergo predictive genetic testing for HD no such medical benefit is created … and thus tests for HD are performed for non-medical reasons.*[134]

However, she argues that:

> *… the existence of medical benefit as an outcome of predictive genetic testing does not negate the potential for harm. It simply overrides this; and the absence of medical benefit does not preclude the possibility of benefit. Predictive genetic testing for FAP is not offered to minors because there is no potential for harm, but rather because the potential for medical benefit is perceived to override the potential for harm. In the case of predictive genetic testing for HD, the absence of medical benefit does not increase the potential for harm, it simply leaves a greater absence of justification for overriding this potential for harm. There is no prima facie reason to assume that the potential for harm should differ greatly between predictive genetic tests provided for FAP and predictive genetic tests provided for HD.*[135]

She therefore does not separate these two types of experiences when conveying the descriptions given to her by the young people that she interviewed.[136]

As a result of her interviews with young people who had undergone predictive genetic testing Duncan provides empirical evidence of many of the purported benefits and

harms referred to earlier. The evidence is from young people who have been through predictive genetic testing, and is not merely conjecture.

*2.5.1.3 Evidence of benefits of predictive genetic testing regardless of test result*

- Enhanced confidence: one young person reported feeling 'nice and important' because his signature was required for the procedure, rather than his mother's;[137] another said that he respected himself a lot more after genetic testing, and realised that he could do anything.[138]
- Self-knowledge gained from results promoted more autonomous decision-making: young people intimated that they had made career plans around the genetic information.[139] Related, Wertz has also written that several young women who had been tested for carrier status for Fragile-X in their teens reported that they were grateful for the test, because results showing that they were carriers helped to not only explain their poor performance in school, but also enabled them to plan their future careers.[140]
- Minors moved through the 'rebellious teenage' stage more quickly, apparently as a result of learning their genetic status, including getting off drugs and getting into less trouble with the police.[141]
- Testing relieved the uncertainty and anxiety of not knowing.[142]
- Testing allowed psychological adjustment at an age-appropriate time. One young woman indicated that she was only fifteen when she was tested and her attitude had been to worry about the here and now, rather than the future: 'at least I know … at least my parents know and don't worry about it'.[143]
- Testing gave minors the opportunity and time to plan their future: after testing, some felt more comfortable talking about the future.[144]
- The testing process improved relationships with family members, and often parents in particular.[145]
- Testing impacted positively on some friendships.[146]
- Testing gave some minors the opportunity to deal with many issues in the genetic counselling sessions.[147]
- The person tested was at an age young enough for it not to have much of an impact:

  *… honestly at 15 I was just such a socialite … I was always out, I was always going to the movies, I wasn't concerned with anything else to be honest.*[148]

#### 2.5.1.4 Evidence of benefits more specifically associated with a mutation-negative result

- Relief in knowing that the genetic condition will not develop:

  *… You can't explain it, just relief, relief … it's like oh my god you know, thank god it's over with, and you've gone this far, and to come this far… A negative result, it's fantastic …*[149]

- Being able to stop worrying.[150]
- Feeling freer, able to plan and excited about the future.[151]
- Positive psychological effects:

  *… it was just grouse … sort of, amazing that you know … I sort of tried to blank it out and then all of a sudden you find out you don't have it … it's so good …*[152]

  *… so excited, I screamed … it was great and I was just dying for this day all my life and it finally arrived …*[153]

  *Life's better … happy and cheery …*[154]

- Feeling that children are a possibility and not having to worry about them developing the condition.[155]

  *We didn't want to have children if they had the gene, ah, cause we didn't want to pass it on…but, yeah, now we do …*[156]

- Allowing parents to stop worrying and seeing parents happy.[157]
- Feeling more able to help gene-positive parents.[158]
- Knowing that the condition stops now in the family.[159]

#### 2.5.1.5 Evidence of benefits more specifically associated with mutation-positive result

- Establishing new relationships with people in similar situations.[160]

#### 2.5.1.6 Evidence of harms regardless of test result

- Testing process can mean time off school and can interfere with school work.[161]
- Anxiety and stress whilst waiting for test results.[162]
- Counselling sessions dragging on: one young person indicated that she had just wanted to be tested, rather than to go through continual counselling sessions.[163]
- Regret about having the test:

  *I would rather of not ever known … cause it's always in the back of your mind, but like, if I hadn't have known, well, what you don't know won't hurt you. …*[164]

- Negative psychological effects, including feeling really lost.[165]
- Wanting more contact with the counsellor than provided after receipt of test result.[166]
- Being too young to understand what the test entailed.[167]
- Feeling distanced from parents who do not share the same genetic status.[168]
- Drifting apart from parents.[169]
- Feeling angry about the manner in which and to whom the results were disclosed, expressed particularly by one young woman who had been told over the telephone that she was positive for the FAP mutation.[170]
- Anxiety about other people learning of the result, and their reactions and potential for gossip.[171]
- Being treated differently and feeling let down by friends who are unsupportive.[172]
- The desire for time alone affecting friendships.[173]

#### *2.5.1.7 Evidence of harms associated with mutation negative result*

- Believing that one's own test result implies a specific result in a sibling:

  *… cause it was 50–50 whether I got it, I was thinking, because I didn't have it, is it more chance [brother] was going to get it and stuff like that …*[174]

- Feeling concerned about affected siblings.[175]
- Feeling concerned about having to look after an affected sibling or parent.[176]
- Feeling guilty about not being affected when siblings or parents are affected.[177]

#### *2.5.1.8 Evidence of harms more specifically associated with a mutation positive result*

- Knowing that the genetic condition could be fatal:

  *[I thought] I'm done in for I s'pose, you know, hitting the ah, hitting the wall now …*[178]

  *… that was … one of the worst days of my life, finding out … because [it] could kill me, and yeah, something I can't get rid of …*[179]

- Negative psychological effects, including increased worry,[180] sadness,[181] anger, and depression:[182] 'At first, I thought, why me and not him … and it's not fair.'[183]
- Watching parents become upset:

  *… um, above everything else, I was hurt when I saw what it did to my parents …*[184]

*When my dad … sat in the bedroom and didn't come out for like 2 hours … when I saw that … that really upset me then … when I saw what it did to my parents …*[185]

- The positive test result resurfacing when other things go badly in life.[186]
- Concern about not being able to perform at work, or being fired.[187]
- One minor indicated that she did not want to identify with sick family members.[188]
- Some minors experienced difficulty integrating with peers because of feeling different.[189]

#### *2.5.1.9 Evidence of harms arising from not testing minors*

- Diminished confidence, competence and autonomy:

  *… the worst thing was getting tested at 18, I felt like I went through 2 years of begging to be tested, 2 years of proving myself worthy enough … proving myself that I'm ok to get tested, that was horrible for me, cause, I'm human, I know that I might have the gene or not, isn't that enough, I actually had to prove myself that I was worthy enough to find out the result, and I really didn't like that …*[190]

- Prolonged the uncertainty and anxiety of not knowing:

  *… for 19 years it feels like I've held my breath …*[191]

- Limited opportunities and time to plan future, including reproductive decisions.[192]
- Prolonged family and friendship tensions.[193]
- Frustration for the minor seeking testing can result in feelings of unworthiness:

  *I rang and she said no, you can only be 18 … I went through this huge anguish, like sort of battle on myself, holding my breath more than ever, um, wondering if I've got this illness or not… You just want to know what way you want to direct your life, you know, how you're going to work around, well, if you don't have it, what you're going to do, if you do have it, what you're going to do, like, it's such a big life choice, you know, and it's sort of a ball, just this, ball game playing with you waiting til you're 18, and I would have loved to get tested at 12 …*

  *I wanted to crawl back up into my mum's vagina just to get tested, you know, that's how desperate I was, I wanted to be a foetus again …*

  *… when you want to know something you want to know, it's like tattslotto … if someone knows what the numbers are, and you don't, you're going to get pretty pissed off with them if they don't tell you …*[194]

- Resulted in or prolonged risk behaviours in minors who knew that they were at risk, including bad behaviour at home and at school, excessive drinking, drug-taking and trouble with the police.[195] The comments from the young people whom Duncan interviewed accord with the findings from Codori et al.'s two studies of the effects of predictive testing for the FAP mutation, in which all minors showed a decrease in behavioural problems following testing.[196]
- Prolonged the period in which minors assumed they had the genetic mutation.[197]

There is evidence that adults too, for whatever reason, assume pre-testing that they have the disorder for which they are at risk:

> *For the last 5 years I kept thinking to myself that I had HC. I thought, 'Yes, you have,' for all the reasons I have laid out before and, in a way, it was easier to cope by convincing myself I had it. It wasn't too hard to do, believe me![198]*

### 2.5.2 Carrier testing

There is a great deal more evidence about the effects of carrier screening programmes than there is in respect of individual carrier testing of people within high-risk families.

It is conceivable that some of the evidence about the effects of carrier screening of minors might be extrapolated to project likely effects of individual carrier testing of at-risk minors. However, note that Michie and Marteau have argued that population-based screening is associated with more problems than screening of high-risk groups.[199]

There is (historical)[200] evidence from carrier screening programmes that knowledge of carrier status can have adverse effects in adults.[201] Clarke provides the following review of the evidence:[202]

- Stigmatisation or fear of stigmatisation.[203]
- Adult Tay-Sachs carriers have been shown to be less optimistic about their future health.[204]
- As many as 19 per cent of Tay-Sachs carriers in one study were worried about their carrier status years after testing.[205]
- 10 per cent of sickle-cell and thalassaemia carriers, in a study in Rochester NY, also worried about their carrier status.[206]
- Wider issues of social stigma have caused concern, particularly in relation to population screening for sickle-cell carrier status in the United States.[207]

- Limited experience with population screening for cystic fibrosis in Britain indicates that many adults, even those related to known carriers, decline the offer of screening tests.[208]

  *The fact that many adults who are offered carrier screening for recessive disorders decline to accept the offer is similar to the low uptake in practice of predictive testing for HD. It is interesting that only 20% of the staff in a medical genetics department in London chose to accept the offer of cystic fibrosis carrier screening, indicating that low uptake rates may be caused by factors other than a lack of knowledge or understanding …*[209]

Wertz reports that early findings by Clow and Scriver about the Montreal Tay-Sachs carrier screening programme (of adolescents aged fifteen and over) were positive: 'These studies suggest that learning that one is a carrier does not ordinarily damage self-image or lead to social stigmatization.'[210]

An eight-year follow-up questionnaire revealed that adolescents screened in the Montreal Tay-Sachs programme were predominantly in favour of having been screened. 'Although 46 per cent of carriers were "worried" after receiving results, the majority were "indifferent" eight years later to the knowledge of being a carrier.'[211]

Further research on the Montreal Tay-Sachs carrier screening programme suggested that many adolescents can effectively use genetic information attained during high school years in later reproductive decisions.

*This analysis shows no negative effects of knowledge of carrier status, and demonstrates that adolescents both remember information regarding their carrier status and use it in an appropriate, mature manner.*[212]

Evaluation of an Australian programme of Tay-Sachs disease and cystic fibrosis carrier screening of 629 students (77 per cent of a possible 817 students) between the ages of fifteen to seventeen in four private Jewish high schools in Sydney in the late 1990s, revealed that:

*Three to six years later, there was a high retention of knowledge, low concern, high levels of satisfaction, and no stigma was experienced by genetic carriers, who reported positive intended result use. … Ninety-one per cent of the sample reported that they were either satisfied or very satisfied with the programme.*[213]

## 2.6 The available evidence from existing research

### *2.6.1 Predictive testing of minors for the FAP mutation*

The evidence from the three studies outlined earlier in relation to predictive testing of minors for the FAP mutation[214] appears encouraging, in that no major harms were associated with testing, even when the test results were mutation positive. Any harmful psychological effects reported were mostly subclinical and were associated largely with minors who had affected mothers or siblings, often regardless of whether or not the minor was actually affected. Thus the issue of whether predictive testing is beneficial or harmful to a minor is not so simple as merely looking at the test results of the individual. The family context, which may be vitally relevant, is discussed in more detail later.

Perhaps the most interesting finding amongst these studies comes from Michie et al. Minors who received a positive test result displayed anxiety and distress levels within the normal range, while 43 per cent of affected adults displayed clinically significant anxiety levels. This finding appears to support the argument that testing during adolescence, at a minor's request, occurs at an age-appropriate time.

The relevance of these studies on testing for the FAP mutation in the context of a competent minor requesting predictive testing is debatable. Many of the children in these research studies were young, and were tested on the basis of parental consent, rather than their own.[215] Codori et al. surveyed children aged six to sixteen in their first study, and children aged five to seventeen in the later study. Parents gave informed consent for their children to participate in the study, and children assented. Michie et al. studied minors aged ten to sixteen, who were tested upon parental consent. The circumstances might be different where a minor seeks and consents to testing, although the findings are encouraging.

Another concern is that these findings in respect of the effects of testing for the FAP mutation may be limited in their application to testing for conditions for which beneficial medical interventions are available. The availability of medical interventions may well have helped the minors adjust to the news that they had tested positive for the mutation (which is not to minimise the seriousness of testing positive for the FAP mutation).

Regardless, testing an at-risk competent minor for the FAP mutation will generally be non-controversial given the medical benefits that accrue from testing: increased surveillance and early detection for those who test positive; and cessation of invasive surveillance for those who test negative.

### 2.6.2 *Predictive testing for HD in adults*

The recorded effects of predictive testing of adults for HD are diverse. There is certainly evidence of harms arising from predictive testing, whether test results are mutation negative or positive. And yet some would argue, as Duncan has, that fears of catastrophic effects as a result of such testing have not materialised.

Notwithstanding, the literature and the evidence of benefits and harms of predictive testing of adults for HD cannot simply be imported into the more general discourse on genetic testing of competent minors upon request, not least because HD is a unique disorder 'terrible in its manifestations, progression and fatal outcome'.[216]

Additionally, there may be many salient differences between competent minors who seek predictive testing for HD and those adults who have been studied in the HD literature. Being adults, and not minors, many of the participants had lived with their at-risk status for a long time, which may have accounted in part for the massive impact experienced even after a mutation-negative result. Also, many of the adults studied had already made marital and reproductive choices and thus the benefits of testing may not have been as immediately obvious for this group. The age of onset was also closer in time for the adults than the minors. The differences between how adults and competent minors react to such testing will remain speculative until more empirical evidence is gathered.

However, evidence exists of benefits arising from predictive testing of adults for HD, even when the results are mutation positive. Of the limited evidence available regarding predictors for reactions to such testing, emotions and other baseline strengths pre-test are important. 'Test result, age and gender have been shown to be bad predictors.'[217]

### 2.6.3 *Duncan's international survey of clinicians*

The responses to Duncan's international survey of professionals involved with predictive genetic testing provided generally positive evidence of outcomes from testing of competent minors: two adverse effects were reported, and nine beneficial effects. However, Duncan rightly notes that there are questions around how to define beneficial and harmful outcomes of genetic testing. It could be argued that the two negative outcomes reported (initial depression and rebellion before eventual acceptance, and concern for affected mother and untested brothers) were entirely normal and appropriate responses, and that the minor's concern for her mother and brothers may well have existed sans testing.[218]

The significant lack of follow-up of the minors tested (only 50 per cent of the younger children tested, and two-thirds of the competent minors, were followed up) is concerning, not only because it limits knowledge of the effects of the testing,[219] but

also because it means that minors and their families may not have been getting the necessary follow-up support.

Duncan also notes that clinicians may have under-reported harmful outcomes of testing because they had 'provided tests, contrary to recommendations made in current guidelines'.[220] Under-reporting of harmful outcomes for this reason may be less likely in respect of competent minors because, as is argued earlier, most of the current guidelines are open to the testing of competent minors upon request.

Finally, the evidence as to outcomes is questionable given that they were reported by the clinicians involved in the testing and not the individuals tested or their families; and there were no common definitions or standard criteria for beneficial and harmful outcomes against which clinicians could measure outcomes. The questions in respect of outcomes simply stated: 'What has the follow-up shown?' and 'Looking back, do you think it would have been beneficial for the young person to wait until the age of 18 years or older to be tested? Why/Why not?'[221]

However, despite the limitations, the results indicate that, amongst the cohort tested and followed up, more benefits than harms accrued as a result of predictive testing.

### *2.6.4 Duncan's research with young people tested for the FAP and HD mutations*

Duncan's study of eighteen young people who had undergone predictive testing for the FAP or HD mutation provides empirical evidence of a litany of benefits and harms that may accrue from predictive genetic testing.

However, the sample size was very small, and involved individuals tested for just two disorders. Additionally, all of those tested for HD (except one) were at least eighteen years of age at the time of testing, and thus the relevance of their responses with regard to the effects of predictive testing of competent 'minors' may be questioned. However, as Clayton and many others have pointed out, minors 'do not emerge from a cocoon at age 18 with full blown decision-making capacity ...'[222] And the young people tested for HD also provide evidence of the largely negative effects on them of being denied testing before the age of eighteen.

In terms of benefits and harms specific to each of the disorders, we recall that Duncan discussed the experiences of both kinds of testing together because of her 'assumption that the existence of a medical benefit would not negate the potential for harm when mature young people undergo predictive genetic tests, just as the absence of a medical benefit would not negate the possibility for benefit'.[223]

However, having analysed the comments made by the young people interviewed, she noted that

> *The experience of predictive genetic testing for FAP is often heavily influenced by the medical surveillance that accompanies a gene-positive test result (or that ceases to be required following a gene-negative test result). Thus, young people's descriptions of their experiences of testing for FAP often focus upon the positive and negative aspects of the provision (or absence) of medical surveillance such as colonoscopies and surgery. … the meaning they ascribe to the test is often centred around this medical outcome. It is therefore fundamentally different from the experiences of young people who undergo tests for non-medical reasons. It is not the range of benefits and harms that alters in these cases, but rather the young people's focus within this range. For example, in the face of a gene-positive test result for FAP, a major benefit articulated by young people was that they would be able to prevent cancer from developing. This, understandably, often overshadowed other benefits such as relief about knowing their genetic status or feeling able to plan life more effectively.*[224]

Duncan therefore cautions that 'we must be mindful of the limitations of using empirical evidence relating to tests provided for medical reasons in order to draw conclusions about tests provided for non-medical reasons'.[225]

Some very positive experiences are revealed in the young people's comments conveyed by Duncan. And despite the evidence of negative feelings also provoked by genetic testing, it seems only one of the eighteen young people interviewed expressed regret about being tested (and he appears to have been tested at the behest of his parents, rather than of his own volition: 'Dad just organised it, and I just had to come in, get the um blood test.')[226]

However, note too the inherent bias (acknowledged by Duncan)[227] that all of the young people consented to be interviewed, and may have been coping better with their genetic testing experience than those who refused to participate in the research. Those who were most harmed by the testing may well have refused to talk about it.

### *2.6.5 Susceptibility testing*

Testing for predisposition to conditions for which the genetic mutation is not 100 per cent penetrant can add a layer of complexity to analysing the benefits and harms. One of the major benefits of predictive genetic testing is the reduction in uncertainty that a test result provides. However, susceptibility tests cannot tell someone whether they will get the disorder tested for, only whether they are at increased risk (or not) for developing the disorder. However, susceptibility tests may reassure those who are found not to have the relevant mutation.[228]

The fact that one is only predisposed as opposed to certain to develop the disorder, and that there are often medical interventions available for multifactorial disorders,

does not necessarily make genetic testing a more palatable experience. Some of the treatments available, such as removal of the bowel and insertion of a colostomy bag, or a double mastectomy or oophorectomy, are far from pleasant.

If one does choose to undergo genetic testing, and tests positive for a genetic mutation that increases one's susceptibility to a disorder, then the test results could herald just the beginning of further difficult decisions. One might consider or undergo extreme and potentially unnecessary measures to reduce susceptibility to the disorder, e.g. a person found to have a BRCA 1 mutation may have a double mastectomy to reduce the chances of developing breast cancer. Being at risk for a mutation that predisposes one to developing a disorder is a drawn out, complex and involved multifactorial issue: not just a 'test or no test' equation.

The possible sequelae and options provoked by a mutation-positive test result need to canvassed before testing is undertaken. The person seeking testing needs to be aware of the implications of the test in order to make a fully informed decision.

### *2.6.6 Research on the effects of carrier testing*

Hogben and Boddington's argument that protection of autonomy is considered to be more pertinent in relation to predictive testing than carrier testing, and that predictive testing of children is thereby seen as more serious because it 'spoils' their autonomy, is not as germane in the context of competent minors who request genetic testing. Genetic testing upon a competent request does not 'spoil' autonomy regardless of the type of test.

While appreciative of the concerns raised by Hogben and Boddington[229] we are unconvinced that carrier testing is not in actual fact 'less serious', in terms of potential harms, than predictive testing for late-onset disorders, not least because of the reproductive implications of predictive testing. People affected by a heritable genetic mutation can also pass the mutation on to their children. Persons found by predictive testing to be susceptible or certain to develop a late-onset disorder may face even greater reproductive challenges than unaffected carriers, given that some are affected by autosomal dominant mutations. This mode of inheritance means that any child of theirs has a 50 per cent chance of inheriting the mutation (and therefore the disorder), regardless of with whom they procreate. Female carriers of X-linked genetic mutations have a similarly high risk of passing on the disorder to any male offspring and carrier status to female offspring. A couple who are both carriers of the same autosomal recessive mutation, on the other hand, will have a 50 per cent risk of passing on carrier status but only a 25 per cent risk of having a child with the disorder in question. Additionally, one has to have a life in order to have reproductive concerns. Predictive genetic tests often test for genetic mutations that threaten life itself.

While some parallels may be drawn between the effects of testing for carrier status as part of a screening programme and testing an at-risk individual on request, there is also need for great caution in comparing the two scenarios.[230] Much of the research on the effects and efficacy of carrier screening programmes focuses on people who underwent such screening more than a decade ago. Genetic knowledge, clinical genetics practice and genetic counselling have evolved since that time.

Even in respect of more recent research into the effects of carrier screening programmes, the correlation is tentative, at best. Participants in carrier screening programmes, and at-risk individuals who seek testing for themselves, may differ in terms of their baseline understanding and emotions, the information that they are given, the opportunity for questions, the availability of genetic counselling before, during and after testing and the handling of test results. There may also be additional pressure to participate in testing when it is part of a programme offered to all of one's peers.[231]

The Australian Law Reform Commission noted that 'the information and counselling needs of screening program participants differ from those who have had experience of a disorder in their families'.[232] Participants in screening programmes may have little understanding of the disorder for which they are being tested, because they may never have met an affected person or had the opportunity to learn about the condition. Added to this, 'the large scale or "production line element" of population screening may place time constraints on obtaining consent and providing counselling'.[233]

However, the fact that carrier screening programmes for certain disorders are seen as relatively common within schools in some communities is arguably an acknowledgement by those communities, at least, that carrier screening of minors is either not significantly harmful, or the potential benefits outweigh the potential harms.[234] Additionally, the education sessions and requisite one-on-one meeting with a counsellor as part of the Australian high school carrier screening programmes appear to have resulted in significant knowledge gains and alleviation of anxiety for the participants in those programmes. And, the evidence from both the Montreal and more recent Australian Tay-Sachs disease screening programmes indicates that carrier screening at high school is beneficial to young people.[235]

## 2.7 The dearth of evidence

There appears to be no evidence for some of the purported benefits and harms recorded in Tables 1 and 2. Indeed, there is evidence contradicting, or at the very least calling into question, some of the purported benefits and harms.

While Duncan's research, for example, revealed that minors did misunderstand aspects of the heritable disorder for which they were at risk when younger (often

when they first became aware of the disorder in a family member), she reported very few misunderstandings post counselling and testing. That is, none of the young people that she interviewed appeared to labour under misconceptions about what the test result meant for their futures.

It has been argued that 'testing during adolescence might interfere with the natural separation process that needs to occur between parents and their children'[236] because of a bond between an affected minor and an affected parent, or because of guilt over not being affected when one's parent is. A range of evidence exists relating to these issues (discussed in greater detail shortly) indicating that the outcome of genetic testing very much depends upon each individual family context. For example, one individual noted that she felt her relationship with her mother had eased somewhat, perhaps because her mother was not so worried anymore:

> *So she sort of let me go off and do what I've gotta do, you know, in my own way, and she knows that's the sort of way I've got to do my thing, like, she can try and drill things into my head over and over again but she knows sort of, she's free to do what she wants now and I don't have to sort of be her carer for life any more, so yeah, if anything we've drifted apart but I mean, our relationship, our friendship's still exactly the same, it's just um, we've drifted apart a bit.*[237]

The purported benefits and harms need to be critically challenged, and not simply accepted, until they are confirmed by evidence.

Some of the purported benefits and harms listed are imported from other contexts, and some could also arise in other scenarios: they are not unique to the genetic testing context. For example, unease about interrupted school work and time out of school may certainly be a relevant concern for competent minors, particularly if they are at an important stage in their school careers. However, having to take time out of school is not unique to the genetic testing scenario – it is an unfortunate potential consequence of any kind of illness experienced during childhood. For older minors, concerns about work and work performance can also occur as a result of many other medical conditions, and they are also perhaps even more of a concern for adults.

Duncan points out that long-term follow-up of minors who have undergone genetic testing is particularly important, because there is evidence from the HD literature that the consequences and value of undergoing predictive testing can change as the time of onset approaches.[238]

## 2.8 Harms apply to adults also

Arguments against genetic testing of competent minors must be examined against the discourse around genetic testing of adults. As Duncan says

> *… it is clear that not all of the arguments used to oppose predictive genetic testing in young people hold the same weight. Most powerful are arguments that are highly specific to the differences that occur between young people and adults (or young people's lives and adult's lives). Less powerful are those that can be applied to both young people and adults.*[239]

Many of the purported and proved harms of genetic testing are just as, and often more, relevant to adults. However, some benefits and harms are more specific to competent minors because of their stage of life, e.g. most are in the process of considering career and other future options, rather than already having made these decisions, and most live in the family home. Certain harms, such as those arising from family relationships, may be exacerbated for young people who live in the family home with their parents and siblings, and who may be financially reliant on their parents. Harms relating to stigmatisation and feeling different may also impact in particular upon teenagers, for whom peer support and acceptance is very important.[240] However, the benefits relating to the ability to make future plans using the test information may be more palpable for minors than adults, given that minors will have greater opportunity to use the information in a timely fashion.

Many of the purported benefits[241] of genetic testing of competent minors upon request may also accrue to adults, e.g. a reduction in risk behaviour, reduced uncertainty and contact with a knowledgeable health professional. However, the benefits of genetic testing for adults will not be discussed further because adults are not the focus of this section of the report, and there is a larger and more relevant body of evidence specifically related to the effects of genetic testing in adults.

## 2.9 General discussion of harms and benefits

The harms and benefits of genetic testing of minors are now discussed without further differentiation between the types of genetic testing undertaken because, as discussed earlier in the context of purported harms and benefits, many of the same benefits and harms apply, to varying degrees, to all kinds of genetic testing (except perhaps symptomatic genetic testing, which more closely parallels general medical testing).

As Duncan has said:

> *It is not the range of benefits and harms that alters in these cases, but rather the young people's focus within this range.*

By this stage, it will hopefully be clear to the reader which benefits and harms may apply more specifically to persons tested for carrier status and which may apply to those tested for a late-onset disorder. The added complexity of susceptibility testing has also been discussed.

## 2.10 Family issues

Family issues are discussed first because the evidence relating to the effects of genetic testing on family relationships is varied and complex, and does not fit neatly into the dichotomy of benefits and harms. Family issues are complex in the context of genetic testing, not least because the genetic information revealed has implications for all family members.

Family relationships and issues also potentially comprise a more salient aspect of the genetic testing process for competent minors for the reasons outlined earlier: competent minors generally live in the family home with their parents and siblings, and are reliant on their parents both financially and emotionally to some degree. How each minor will react to the vagaries within the family context as result of genetic testing is far from clear. There is great speculation about whether genetic testing strengthens or strains family relationships. There is limited and contestable evidence for both propositions.

No evidence has been found by this author to suggest that a competent minor who requests a genetic test and is found to be mutation positive is likely to be scapegoated or treated poorly within the family because the mother or father are reminded of their own 'unacceptable traits'.[242] That of course is not to say that this would never occur, but evidence for the proposition is currently limited in this specific context.

It has been suggested that an affected parent may not be able to provide appropriate support for a minor who tests positive for the genetic mutation, because of the parent's own struggle with the condition. Evidence directly on point is also lacking for this proposition. Additionally, a lack of parental support for a minor facing current or future health issues can equally occur in other contexts, and is not unique to genetic testing. It is an issue that needs to be considered in pre-test genetic counselling, to enable the minor to identify and establish other support networks. We reiterate, however, that the evidence is uncertain. Indeed, there is evidence that returning a mutation-positive test result actually creates or strengthens bonds between affected parents and children.[243]

Some minors who test mutation positive may especially suffer while watching a symptomatic parent struggle with the disorder, deteriorate or even die. The strength of this purported harm of predictive genetic testing is weakened by its applicability to adult children also. However, a minor who lives with a symptomatic parent would be

exposed to that parent on a more regular basis. On the flipside, it may help the minor to confront the reality of the situation, and even inspire strength. It may very much depend upon how the parent and the rest of the family responds to the disorder. This is another issue that needs to be thoroughly explored and prepared for in genetic counselling. Note that unaffected minors and indeed any minors with ill parents will also be affected by watching a symptomatic parent struggle with a condition.

There is evidence that minors are negatively affected by mutation-positive results in parents and siblings, regardless of their own test result. There is also evidence of slightly increased anxiety if a minor's mother is positive for FAP and so is the minor; conversely, there is evidence of negative emotions such as guilt and worry for mutation-positive siblings when the minor is mutation negative.[244] In the context of these pieces of research, the anxiety levels of control groups of at-risk minors not tested at all, or minors not at risk, might have been enlightening. One would expect some degree of anxiety among children with ill parents or siblings, regardless of whether or not they are themselves at risk for a similar or different condition.

These results suggest that minors are likely to be troubled by family members' conditions regardless of whether or not they are personally affected or have even been tested, or whether or not the affected family member is suffering from a heritable genetic illness or some other affliction. The possibility of negative feelings arising from test results of other family members is not a sufficient argument against genetic testing of minors upon request; it is something that must be managed.

The impact of genetic testing upon family relationships is much more complex and contextualised than is suggested by propositions such as testing may 'interfere with the natural separation process', 'drive a wedge or distance between family members' or 'force minors into the realization that their parents are not all-powerful'. Certainly minors' feelings about genetic testing and test results seem to relate to their family relationships and other family members' risk status in particular.

The young people that Duncan interviewed reported feeling concerned about their parents' reactions to their test results. Some who tested mutation negative said that the happiness of their parents was the best thing about being tested, while others who tested mutation positive said that 'seeing what it did to my parents'[245] was the worst thing about being tested.

There is also evidence that testing did result in negative family issues for some minors. One young woman found to be mutation negative for FAP, for example, perceived her affected mother to be jealous of her.[246] Some young people who tested mutation positive admitted to feeling guilty or anxious that their siblings were affected.[247]

Yet many of the young people that Duncan interviewed reported that they had grown closer to family members, usually an affected parent, as a result of the testing process and the test result.[248]

What is clear, from the limited evidence available about predictive genetic testing of minors, is that genetic testing is not undertaken in a vacuum: minors are aware of the impact of the genetic disorder, the testing process and the test results upon their family members (both present and future), and others around them. These issues must be explored during genetic counselling.

There are as many unique family situations and relationships as there are families. For this reason, the potential benefits and harms for a particular competent minor seeking genetic testing can only be accurately assessed on an individual basis and generally against the backdrop of the family relationships. There may be further complexities where the minor does not tell the parents about seeking testing, or does not have their support for testing. Family counselling may be appropriate where all members are amenable.

If these mixed reports tell us anything about how genetic testing can affect family relationships, it is that every individual and every family is unique and the minor's family situation must be carefully explored in genetic counselling pre-testing.

## 2.11 Harms

Many of the harms that may arise in the context of genetic testing of competent minors, upon request, may also arise in the context of genetic testing of adults. Additionally, many of the harms are not exclusive to genetic testing situations, and others are not intrinsic and can be mitigated or managed.

### *2.11.1 Negative psychological effects*

There is certainly evidence of negative psychological effects resulting from genetic testing, both in minors and adults. Indeed, it is arguable, based on the limited evidence available, that predictive genetic testing may be more harmful psychologically for adults than for minors, given Michie et al.'s findings in respect of minors and adults tested for the FAP mutation.[249] Adults in most cases have lived with the knowledge of being at risk for a longer period, with all that entails. Being at risk for the condition may constitute a much greater part of the adults' identity and life story; they will most likely have made more life choices based upon their at-risk status; and there is evidence that at risk people often live as if they were mutation positive. Finding out that one was mutation negative, after having made significant sacrifices, perhaps involving career, marital or reproductive choices, on the assumption that one was affected, could clearly be devastating.

Some negative feelings as a consequence of genetic testing are almost inevitable, particularly when one tests positive for a life-threatening disorder. The probability of feeling some negative psychological effects needs to be fully explored in genetic counselling before testing. Counselling needs to be available to follow up minors who receive mutation negative results also, because there is evidence that people experience negative psychological effects even after receiving a 'good' result.

The severity and duration of psychological harm caused by genetic testing may vary according to the type of testing, the penetrance of the condition tested for and the medical interventions available, if any. However, there is little evidence comparing the effects of genetic testing for one kind of disorder versus the effects of testing for another disorder.

The one young person that Duncan interviewed who expressed regret at undergoing a predictive test had tested positive for the FAP mutation.[250] Interestingly he said 'what you don't know won't hurt you', when in actual fact there are medical benefits available for carriers of the FAP mutation. That same young man appeared to have been tested at the behest of his parents, rather than of his own volition: 'Dad just organised it, and I just had to come in, get the um blood test'.[251]

The likelihood of regret might be less of a concern when one is tested of one's own volition. The possibility of regret can hopefully be minimised by full exploration of the possibilities and consequences before one decides whether or not to be tested. However, it may prove impossible to entirely eliminate the possibility of regret regarding the seeking of information that one does not want to hear.

Undoubtedly negative psychological effects frequently occur for *anyone* who discovers that they have a debilitating or life-threatening disorder. Such feelings are not the exclusive domain of those who have undergone predictive testing for a disorder.

#### *2.11.2 Establishing an identity might be difficult*

To date there appears to be no evidence that competent minors who seek genetic testing have difficulty establishing an identity if they test positive for the disorder. The suggestion that one might have difficulty establishing an identity seems to be more specifically related to minors than adults, given the period of growth and development young people go through. However, note the concern raised earlier that adults have lived at risk for longer and thus may be equally if not more likely to experience an identity crisis upon receiving their test result. The suggestion also appears to ignore the fact that the minor is already living as an at-risk individual in an affected family, and is not a *tabula rasa* upon which the test results are laid.[252]

Genetic counselling can help both minors and adults incorporate their genetic status and test results into their identities.

### *2.11.3 Perception that a mutation-positive minor is already ill*

The concern that people, particularly parents, will treat an individual found mutation positive for a late-onset disorder as if they were already ill is a concern more frequently raised in the context of genetic testing of younger minors at their parents' request. However, it is also a potential harm that could be faced by anyone who tests positive for a late-onset disorder, including adults.

Again, the argument fails to consider the baseline harms of simply being at risk for a condition. Minors, and others, may already be treated as if they have the condition and are 'ill' if they are at risk, regardless of whether or not they undertake predictive testing. Indeed, it has been suggested that some family members may be singled out as being 'affected' within a family, without testing, perhaps because their health is generally more fragile, or they most closely physically resemble the affected parent.[253]

Appropriate counselling before the decision is made to undergo testing, during the testing process and after testing should aim to curb this type of misunderstanding and misuse of information: the potential harm is an information management problem that can be addressed.

### *2.11.4 Concerns about disclosure, integration and stigmatisation*

Concerns about disclosure are also relevant to adults – they are not unique to young people. Where confidentiality requirements and rules are adhered to and correct processes are followed the disclosure of test results to others should not be an issue (except where disclosure is required by law). Minors who have given their own competent informed consent to genetic testing are also entitled to confidentiality: their results should not be disclosed to anyone else, including their parents, except where one or more lawful exceptions applies. This is discussed in more detail later, under the heading 'Privacy'.

Competent minors may be particularly vulnerable to anxiety about integrating with their peers and being stigmatised if their test results are known, given the social pressure to conform and 'fit in' during adolescence.[254] However, disclosure of information can and should be managed. Fear about inappropriate disclosure is not a reason to prohibit genetic testing of minors: it is an impetus to safeguard confidential information. The person tested should be in control of the information. Managing the information and deciding when to tell others, including potential sexual partners, is an issue that needs to be covered in genetic counselling of minors.

### *2.11.5 Being too young to understand*

The potential harm of being too young to understand the implications of the genetic test should *never* be an issue if the competency assessment of the minor requesting testing is appropriately managed. The focus of this section is on genetic testing of *competent* minors upon request.

## 2.12 Benefits

### *2.12.1 Enhanced autonomy, competence and confidence*

There is explicit evidence that undertaking genetic testing on the basis of the minors' own authority enhances their confidence: '… it required my signature, not mum's, I remember that which was of course quite surprising being under 18, it actually made [me feel] quite good and nice and important'.[255]

There is also evidence that decision-making more generally enhances minors' competence: 'research has long-since confirmed the common-sense belief that children need to exercise choice in order to acquire self-respect and a sense of responsibility'.[256]

### *2.12.2 Self-knowledge promotes more autonomous decision-making*

A number of the young people whom Duncan interviewed indicated that they had readjusted their career and reproductive expectations because of the genetic information and self-knowledge gained. Indeed, the concept that self-knowledge promotes more autonomous decision-making is arguably an *a priori* proposition, depending upon one's definition of autonomy.

> *Autonomy is self-government or self-determination. Being autonomous involves freely and actively making one's own evaluative choices about how one's life should go. … Autonomy requires some concept of self and self-knowledge. Autonomy, at least in the Millian sense, is related to forming one's own conception of the good life for oneself, and acting on it. But to decide what the good life is for oneself, it is necessary to know what kind of entity one is. A fundamentally important fact about ourselves is how long we will live and how robust our health will be. To take the extreme case, it might make a great difference to our actions, if we learnt we were to live one more day or 40 more years.*[257]

Even if one accepts that increased self-knowledge enhances autonomy, one need not accept that predictive testing of minors always benefits their autonomy. The benefit of self-knowledge to the minor's autonomy arguably only accrues when *choice* can be exercised regarding whether or not to acquire more information, including genetic information. In this way, predictive testing of minors who cannot consent or who have not consented on their own behalf does not necessarily enhance their autonomy by giving them more self-knowledge upon which to make future decisions. And yet, self-knowledge is an added benefit of being able to exercise one's own choice in respect of testing.

### *2.12.3 The impact of genetic testing on minors' behaviour*

There is limited evidence as to how genetic testing affects minors' behaviour. Thus far there appears to be little evidence that genetic testing of minors causes behavioural

problems, and yet there is quite a body of evidence (relative to the limited total amount of available evidence) to indicate that genetic testing can impact positively on problem behaviour. Personal accounts from the young people that Duncan interviewed indicate that some of them indulged in serious risk-taking before being tested, including drug abuse and criminal or disorderly behaviour. Some felt that having the genetic information influenced their risk-taking behaviour positively.[258] Codori's research into the effects of testing for FAP also indicates that predictive testing was not associated with an increase in behavioural problems.[259] The limited evidence suggests that genetic testing of minors, at their request, might impact more positively than negatively on their behaviour.

#### *2.12.4 Testing relieves uncertainty*

Relief of uncertainty is another effect of genetic testing, which is arguably knowable *a priori* (to the extent that a useful result is produced by the test). There is also evidence of the relief experienced by young people who have been tested, as outlined in Duncan's research. However, the evidence is certainly limited (see the earlier discussion on Duncan's research), and there has seemingly been no longer-term follow-up on the value of reduced uncertainty for minors versus the knowledge that one has or is predisposed to a potentially terminal condition.

#### *2.12.5 Direct contact with a knowledgeable health professional*

Direct contact with and counselling from a health professional with a good understanding of the clinical and ethical aspects of genetic testing can be of major benefit to a minor (and others). Up until contact with such a health professional the minor may have gained all information and ideas about the disorder from within the family, directly or indirectly. Contact with a knowledgeable health professional can be extremely beneficial in that context, regardless of whether or not testing goes ahead.

#### *2.12.6 More accurate genetic counselling becomes possible*

More accurate genetic counselling is also a self-evident benefit (where the results of the test are not inconclusive). A minor attending genetic counselling to discuss and learn about the disorder and the implications of the disorder will enjoy more accurate counselling about the risk and the condition, no matter whether a mutation-positive or mutation-negative result is received. After testing the minor can be counselled as to the implications of the result. For example, counselling for those who receive a mutation-negative result may focus on the fact that it is normal to experience negative feelings after a 'good' result, and how to deal with family issues, including family members who are mutation positive. Counselling for those who test mutation positive, on the other hand, can assist them to adjust to the information, explore the implications and plan for the future.

### *2.12.7 Undergoing testing at an age-appropriate time*

There is some evidence that the young people that Duncan interviewed felt that the potentially negative or fearful features of going through predictive genetic testing were mitigated because of their age:

> *… it crossed my mind and I'd think about it a little bit but honestly at 15 I was just such a socialite … I wasn't concerned with anything else to be honest.*[260]

> *If I had of had the test done now, it would have had a bigger impact on me than what it would have six years ago because I think if you are younger you don't really sort of have the same sort of knowledge that you have when you're older and things can have a bigger impact on you.*[261]

However, to the extent that minors were not as worried because they did not understand the implications of the genetic test is not an argument in favour of testing *competent* minors upon request. This argument could be used to justify genetic testing of young children who cannot consent on their own behalf, which is dealt with elsewhere in this report.

However, as highlighted earlier, Michie et al. found more negative effects in terms of increased anxiety levels associated with pre-symptomatic testing for the FAP mutation amongst adults than amongst minors. Suggestions that the difference in anxiety levels between children and adults were due to the minors' lack of understanding regarding the test results or their implications were not supported by the data (see earlier discussion).[262]

### *2.12.8 Time and opportunity to plan future; time for adjustment and research*

One of the major arguments in favour of testing competent minors upon request (and related to the earlier point) is that testing at such a time gives the minor greater opportunity and time to plan for the future around the information revealed. For many minors, choosing subjects to study towards a career begins in high school.

The benefit of having time for adjustment and research is also related to the earlier points. One woman recalled being caught 'off-guard' by her own breast cancer diagnosis and having felt rushed into making uninformed decisions. She considered that a positive genetic test result would have allowed her time for research and adjustment and in turn more informed decision-making. 'Time to make good decisions and to get information gathered. … when I got my diagnosis, I didn't feel like I had the time … I didn't take time to do the research because I wasn't emotionally ready to do that.'[263]

A young woman who knew she had a BRCA 1 mutation would have more time to digest the information. She might 'decide against a long university education to have children early and pursue a university career after having had bilateral mastectomy',[264] rather than being taken by surprise and rushing into a mastectomy.

### *2.12.9 Friendship issues*

How the test result will affect their friendships and other peer relationships might be something that minors are particularly concerned about. In terms of the reactions of friends to the genetic testing process, there is less available evidence than there is for family reactions. However, the little evidence that exists (from the comments made to Duncan) suggests that, like family relationships and reactions, whether testing will impact negatively or positively on friendships appears to depend upon the individual's circumstances and friends. At least two young people, for example, told Duncan that friendships had flourished as a result of the genetic testing process,[265] while others indicated that they had been let down by people, or did not get as much support as they needed.[266]

Minors should be able to control their genetic information and thus restrict their friends' knowledge of their genetic status if they wish. Special genetic counselling around how to deal with friends' and peers' questions and reactions may be particularly important for minors, if this is likely to be an issue.

### *2.12.10 Positive psychological effects of receiving a mutation-negative result*

Duncan reports numerous positive psychological effects, as a result of mutation-negative results, from her interviews with minors who had been through the testing process. The relief, joy and excitement are palpable from their comments.

### *2.12.11 No longer being treated as if one has the disorder*

The concern that people who are at risk for a heritable disorder may be treated by family members or others as if they already have the disorder is discussed earlier. Returning a mutation-negative result would hopefully quell this kind of behaviour.

However, cessation of this kind of treatment should not need to be considered as a 'benefit' of a mutation-negative genetic test result. A person known (by whatever means) to be at risk for a heritable disorder should not be treated as affected until that person confirms the information (and obviously, at that point, there are also anti-discrimination protections.) This is a concern relevant to adults too (perhaps even more so) and it involves much wider privacy and discrimination issues than can be adequately addressed here.

## 2.13 Magnitude of benefits and harms

The probability of occurrence of any of the benefits and harms discussed, and their magnitude, will depend upon the individual and support networks. As stated earlier, in the discussion about predictors of reactions to predictive HD testing, individuals' negative feelings prior to testing (e.g. avoidance of HD-related situations and dissatisfaction) were often exacerbated post testing (e.g. avoidance behaviour,

depression, suicidal ideation).[267] The magnitude of benefits and harms may further depend on the test result; the medical interventions available, if any; the probability of developing the disorder; and the likely age of onset of the disorder.

### 2.14 The impact of the test result

Many of the purported non-medical benefits and some of the purported harms may accrue regardless of whether the genetic test results are positive for the genetic mutation, or mutation negative. It is not only those who receive a mutation-negative result who might suffer some harm. Certainly there is evidence of non-medical benefits accruing to minors who receive a mutation-positive result, as well as to those who receive a mutation-negative result. Thus, anxious desire for a mutation-negative result is not the only reason for seeking a genetic test, and a mutation-negative result is not the only outcome that may produce benefits.

However, the possibility of a minor returning a mutation-negative result commonly appears to be overlooked in much of the literature, which seems to focus on the worst-case scenario.[268]

### 2.15 The importance of an individual baseline assessment before genetic testing and the harms of not testing

Duncan criticises commentary on genetic testing of minors which appears to assume that a minor, or any individual, comes to testing as a blank slate, i.e. an empty page upon which the results can be imposed and which can be utilised to predict the effects of testing.[269]

For example, simply accepting at face value that interrupted school time and school work may be harmful by-products of genetic testing denies recognition that at-risk minors may be having difficulties at school and in other areas of their lives anyway, including difficulty concentrating or a tendency to act up. Duncan's interviews with young people revealed that a number of them were engaging in risk behaviours pre-testing.[270]

An implicit assumption that minors are blank slates pre-test may be seen in the suggestion that there is no harm in testing being delayed until the minor reaches the legal age of majority or the commonly and arbitrarily mooted age of eighteen. Duncan's interviews with minors who had lived with the knowledge of their at-risk status for years before being tested suggest that there may be harm in making a minor wait until the age of eighteen to be tested. The reasons why the minor is seeking the test may be particularly insightful in this regard.

The evidence outlined earlier, particularly from Duncan's study, reveals that many minors (and adults) convince themselves that they are mutation positive before or

without being tested, seemingly to prepare themselves for the worst. Thus, some of the harms of genetic testing may already apply to minors who are living as if they are mutation positive. For example, they may be making career, relationship or other decisions or sacrifices based upon a false idea of their future health.

Not all commentators suggest that people are blank slates pre-testing. Fanos is one commentator who suggests that, rather than being a blank slate, minors who have grown up in families with affected persons may be more affected by genetic testing because 'the impact of a testing procedure will be overlaid upon the pre-existing stresses and adaptations of the child who may be at risk for the genetic disorder'.[271]

A better approach might be to consider the minor and the situation as unique, and assess the likely effects of testing without making assumptions that the minor is a blank slate entirely unaffected by being at-risk, or is more or less prepared for testing than others, who are not significantly at risk, might be.

In support of an individual assessment of each case, Sharpe argues:

> *If this position is adopted as standard policy for all children, does this not in its practical effect reduce the child, and the parents to an abstract generalization, medical genetics decision making undertaken not in response to the individual parent's needs, values, objectives [Ad Hoc Committee, 1975], and right of personal autonomy [Fletcher et al., 1985], but rather upon preconceived assumptions and values that may prove invalid on a case by case basis? … Has the geneticist effectively imposed upon others his/her own values and beliefs about what is 'best' for the child and the family [Shaw, 1987]?*[272]

## 2.16 Conclusion

Empirical evidence of benefits and harms of genetic testing is very limited. However, the most recent and extensive evidence points towards testing having the potential to be more beneficial than harmful for competent minors who request testing.

For some of the purported benefits and harms there is no evidence, or inadequate evidence only. Other purported harms do not sufficiently justify a conclusion against genetic testing of competent minors upon request because they relate equally to other health-care contexts; they relate to adults also; or they can be mitigated or resolved via alternative methods as opposed to blanket prohibition.

Many of the potential harms should not be an issue if correct processes are adhered to, particularly around clear procedures and timeframes for counselling and testing; and clear rules and procedures regarding method, timing and persons to whom disclosure of results will be made.

Some of the benefits of genetic testing are knowable *a priori* and, at the time of writing, there appears to be more convincing evidence for the *benefits* that arise from genetic testing than for the harms, particularly for competent minors. Evidence of harms is limited and harms specifically related to competent minors are even more speculative. Additionally, some of the harms projected as a result of genetic testing can already be seen in the context of minors living at risk for a heritable genetic mutation.

There is a great need for thorough genetic counselling before a decision is made about whether to undergo genetic testing. The benefits and harms of testing frequently depend on the individual's life story and relationships prior to testing, and plans and level of support after testing (particularly where other harms relating to information management have been dealt with appropriately). Evidence from predictive testing for HD in adults shows that predictors of negative psychological effects post testing included negative feelings in the pre-test period.[273]

> *In conclusion, the issue is complex and troubling. It illustrates the importance of considering each patient as an individual, with particular needs, values and objectives: that a decision to administer or nor to administer the procedure must be determined on a case by case basis, and not as result of predetermined values, objectives, and policy.*[274]

Given the absence of convincing evidence that genetic testing is too harmful for minors generally, competent minors should not be denied the opportunity to undergo genetic testing at their request. The next section will address whether the law currently permits competent minors to consent or refuse to consent to genetic testing.

> *Although acknowledging that empirical research can have an important role in developing policy guidelines and in affecting ethical decision-making processes, we stress that the results from empirical research alone cannot determine what is good or bad, right, or wrong. ... Ethically salient issues, such as maintaining the child's right for autonomy, confidentiality, and privacy, must be primarily considered when developing formal guidelines …*[275]

## 3 MINORS WHO CANNOT GIVE INFORMED CONSENT TO GENETIC TESTING

> *… it is clear that predictive testing in childhood for late onset disorders, even in the context of a high risk family situation, can raise as many problems and as much anxiety as is generated by continuing anxiety about the child's genetic status, and the knowledge that a child will develop such a disorder may, at least in some family contexts, cause worse problems than continued uncertainty. At present, we have no means of identifying those families that would be helped by having uncertainty resolved by genetic tests and distinguishing them from other families in which the results of testing would be harmful. Indeed, there is not even any agreed means of deciding whether or not a family might have been helped or harmed by such an intervention, and what time scale should be considered in coming to a judgement on this.*[276]

### 3.1 Purported benefits and harms

The few genetic cancer predisposition syndromes for which genetic test results can alter clinical management in children include: FAP; multiple endocrine neoplasia, types 1 and 2 (consisting of autosomal dominant mutations in genes regulating cell growth, characterised by tumours in the endocrine glands); Von Hippel-Lindau (autosomal, dominantly inherited multisystem disorder characterised by abnormal growth of blood vessels);[277] retinoblastoma (childhood cancer arising from immature retinal cells in one or both eyes);[278] and neurofibromatosis, type 2 (characterised by multiple tumours on the cranial and spinal nerves).[279,280]

What about the non-medical benefits regarding genetic testing of children? Interpretations of best interests or benefit are integral to any debate around genetic testing of children. For example, some may argue that carrier testing or predictive genetic testing, even for an untreatable disorder, is in a child's best interests because:

- It alleviates anxiety for both child and parent: a less anxious parent makes for a less anxious child. If the results are negative then much of the anxiety can fall away immediately and, even if the results indicate the genetic mutation being tested for is present, the certainty may alleviate some anxiety.
- It facilitates open relationships within the family and relevant community.
- It might reveal that the child is mutation negative.
- It gives a child time to adjust to the prospect of life with the condition.
- It allows the parents to secure the best environment that they can for the child, e.g. a house with suitable access that is near a hospital and appropriate school.[281]

Opponents of genetic testing of children argue that carrier testing and predictive genetic testing are not in a child's best interests, for a variety of reasons:

- Development of a perception that the child is 'ill' with attendant negative parental (and other) attitudes towards the child.
- Strict health (including dietary and environmental) regimes and treatments may be implemented with few if any proven benefits.
- Low self-esteem on the part of the child.
- Serious psychological maladjustment, perhaps leading to depression and even suicide.
- Parental guilt, impacting negatively on the child.
- Social discrimination, e.g. in employment and access to insurance.
- Violation of the child's future autonomy and 'right not to know'.[282]

These benefits and harms are similar to those postulated by the professional position statements and guidelines discussed earlier, and reinforce the comments made by various groups which were surveyed regarding their attitudes towards genetic testing of children. Unsurprisingly, they are also similar to the putative benefits and harms of genetic testing of competent minors.

Much of the discussion in those documents and in the preceding section on competent minors therefore also applies to requests for carrier or predictive testing of children who cannot give their own consent. This section focuses specifically on the different issues raised by genetic testing of children who cannot give informed consent (compared to testing of those who can).

Generally, the harms mentioned in respect of genetic testing of incompetent minors are variations on the same themes: violation of the minor's right to autonomous decision-making; and potential psychosocial harms as a result of testing in childhood. Arguments centre around the lack of choice for the minor, and violation of the right to 'not know' genetic status, the lack of confidentiality afforded to minors who undergo genetic testing in childhood and the psychosocial harms that may eventuate within the family and wider social context as a result of testing.

That parental requests may dictate the services made available seems potentially of concern since parents may not always act in the interests of the specific child; rather, they may have their own interests, or the interests of other family members, in mind.

Some commentators argue that parents have a right to consent to genetic testing of their children because they know their children best and they bear primary responsibility for them.[283] Others argue that parents need to exercise significant discretion in raising

their children, and that society has an interest in protecting the family as the primary child-rearing institution.[284] Allowing health professionals to regulate parents' access to medical procedures for their children is therefore paternalistic.

To draw out the issues, Wright Clayton hypothesises about the potential sequelae to predictive testing of a child for a later onset condition (in this case, breast cancer):

> *Hopefully, the parents would help their children to prepare for the surveillance she will need and the possibility that she might develop cancer. However, the parents might discourage her from getting married or having children, since she might not live 'long enough' or she might pass the mutation on to her children. They might decide against sending her to college or professional school because she might not get enough 'good' out of a costly education.*
>
> *Many people would agree that the latter courses of action would be unfortunate, if not misguided for any number of reasons. First, inherent in the nature of predisposing mutations is the fact that the daughter may never get sick. … Second, causing her to feel like 'damaged goods' who should eschew important human relationships and not try to maximize her potential seems harmful even if she ultimately develops cancer. Third, the cancer may not be fatal.*[285]

### 3.2 Lack of evidence

Although there is more evidence on the effects of predictive or carrier testing in adults, children are not small adults: they have their own set of interests, vulnerabilities, capacities and needs.[286] There is a dearth of evidence regarding the effects of predictive testing of children for early or late-onset conditions. The limited evidence that exists indicates that there may be some benefits – or a lack of significant harm – as a result of testing. However, long-term studies may be needed to reach a valid conclusion on the balance between benefit and harm.

Research into predictive testing of minors for the FAP mutation comprises the largest body of evidence relating to the effects of predictive testing of minors.[287] Most of the children tested for FAP (involved in the research presented earlier) were tested on the basis of parental consent, rather than their own. Codori et al. surveyed children aged six to sixteen in their first study, and children aged five to seventeen in the later study. Parents gave informed consent for their children to participate in the study, and children assented.[288] Michie et al. studied minors aged ten to sixteen, who were tested upon parental consent.[289]

Comments from many of the young people tested for FAP and interviewed by Duncan also indicate that they had not been tested of their own volition, e.g.

> *Mum and dad, they decided that they wanted to just see if we had it or not…*
>
> *…when I got to like, I think I was 15, they said oh it's probably time to bring Ali in now …*
>
> *I was 12 when I was told that I had to have the test … I didn't want to have it, but then I sort of had to …*
>
> *Mum was told by the doctors I think, that we needed to get a genetic testing …*
>
> *Dad just organised it, and I just had to come in, get the um blood test …*
>
> *They didn't ask us do you want to, they said you know you have to go get a blood test …*[290]

The evidence from the three studies outlined earlier in relation to predictive testing of minors for the FAP mutation,[291] and from Duncan's research, appears encouraging, in that no major harms were associated with testing, even when minors tested mutation positive. Any reported negative psychological effects were mostly subclinical and were associated largely with minors who had affected mothers or siblings.

Minors who received a positive test result during Michie et al.'s study displayed anxiety and distress levels within the normal range, while 43 per cent of affected adults displayed clinically significant anxiety levels.

The limited evidence available supports the appropriateness of such testing. However, these findings may be limited to testing for conditions for which there are beneficial medical interventions available. The availability of medical interventions may have helped the minors cope with the information that they had tested mutation positive.

In terms of benefits recorded as a result of predictive testing for HD in young people (e.g. those young people tested for HD by Duncan), even greater caution is required if inferring that similar benefits may accrue to children tested for untreatable disorders on the basis of parental consent, rather than their own. Much of the emphasis in the discussion on competent minors focuses on the benefits of predictive genetic testing that may accrue explicitly as a result of testing being undertaken pursuant to the minor's own autonomous choice.

Additionally, predictors of outcomes identified by the literature on predictive testing for HD in adults cannot be similarly applied with respect to minors who cannot give their own informed consent to genetic testing. For younger children it will not be possible to evaluate ego strength, depression levels and coping strategies pre-testing, making it even more difficult to predict the effects of testing on them than on competent minors or adults.

There is a lack of evidence of benefits of childhood genetic testing for untreatable late-onset disorders; there is also a lack of evidence of harm caused by such testing. However, there are two other major arguments opposing genetic testing in children where it is not medically indicated: testing upon the basis of parental consent violates the minor's autonomy and right not to know the information; and such testing results in a potentially detrimental breach of confidentiality regarding the minor's genetic information.

### 3.3 Violation of autonomy

It is argued that carrier or predictive genetic testing in childhood breaches a child's future autonomy by removing the right to make a decision about testing as an autonomous adult or competent minor. Such testing precludes the possibility that, as an adult, the individual might have preferred to live with uncertainty instead of certain knowledge of the mutation.[292]

However, it has been argued in response that parents make many decisions to try and 'shape their children's futures in ways that both broaden and narrow the children's options'. Many states (including our own) grant parents much authority and leeway in terms of how they raise their children, and only intervene when there is evidence a child might be significantly harmed by a parental decision or action (or omission). There is as yet insufficient evidence that predictive testing for late-onset conditions harms children or adolescents so as to warrant state intervention.[293] Equally, however, there is insufficient evidence of benefit, so that – except in cases where benefit can derive from the test – there seems no reason to permit parents to consent on their child's behalf.

Robertson and Savulescu have intimated that the claim that predictive testing infringes a child's future autonomy rests upon the assumption that autonomy or liberty equates with having as many options open as possible. They argue, however, that more choice is not necessarily better; and that childhood testing does not result in *reduced* options, just *different* options (e.g. growing up with information about the future). They also argue that genetic knowledge may in fact promote autonomy by enhancing the subject's concept of self and self-knowledge, which in turn is useful for forming one's own conception of the good life for oneself.'[294]

However, it is often cited that just 10–15 per cent of adults at risk for Huntington disease opt for testing to discover whether they have the genetic mutation,[295] indicating that, for some untreatable disorders at least, most adults exercise their autonomous choice in favour of not knowing. On this evidence is it fair to make that kind of decision for a child, when it is possible to delay such a decision indefinitely or permanently, and at least until adulthood?[296]

A major reason for emphasising a child's right to autonomy or future autonomy in the genetic testing context is the concern to preserve the 'right not to know' whether one carries a genetic mutation, and all of the accompanying sequelae. Hogben and Boddington contend that arguments relying on the principle of respect for autonomy are often considered to be more important in relation to predictive testing. In contrast, knowledge of carrier status is often presented as if it is something to be used or not: as pragmatic information, primarily important for possible action rather than as significant knowledge *per se*. This point has particular salience in the debate regarding genetic testing of younger children:

> *In relation to predictive testing, implicitly autonomy is seen as control over self-image or self-knowledge, which is ipso facto spoiled by revelation of disease status. In contrast, with regard to carrier testing, autonomy is implicitly seen primarily as requiring control over actions. In such cases, then, despite imposing knowledge of carrier status on a child, it can still be claimed that autonomy is maintained as when the time comes, the child can 'choose' whether or not to use the knowledge.*[297]

The concern is that these assumptions tend to 'ground conclusions regarding predictive testing in the relatively absolute notion of autonomy, and conclusions about carrier testing on the more contextualized notions of harm and benefit'.[298]

### 3.4 Privacy and confidentiality

The protection of a minor's autonomy when decision-making about genetic testing is connected with protection of privacy. A child who undergoes a genetic test in response to a parental request will generally not be granted the same confidentiality as an adult, given that the results will be disclosed to the parents, who are not bound by any legally recognisable duty to keep the information private. This issue and its implications are discussed in greater detail in the section on 'Disclosure, confidentiality and privacy'. Minors who undergo genetic testing as children cannot deal with their genetic information as they see fit or keep it private should they wish to do so.

### 3.5 Predictive testing for untreatable early onset conditions

Predictive genetic testing for an early onset condition for which no beneficial medical interventions exist raises fewer concerns than testing for a similar late-onset condition. The same potential benefits exist, but not the same harms. The danger to the minor's future autonomy to make the decision and potentially to exercise the right not to know the information is not as salient, as the child may never reach an age at which to decide whether or not to undergo predictive testing for the disorder in question – having already developed it, or having passed the likely age of onset unaffected.

Thus, the putative benefits of such testing (such as relieving anxiety, preparing for onset) may be weighted more heavily in this context regardless of whether or not the disorder is treatable.

However, given that there are no clinical benefits to such testing, and there may be some harms (such as changed parental expectations and treatment of the child), parental requests for such testing should still be treated very cautiously.

In terms of genetic testing providing some certainty, or alleviating anxiety, there is a great deal of residual uncertainty regarding the interpretation of predictive genetic test results. When genetic mutations are not fully penetrant, and only indicate susceptibility to a disorder, the risk of developing the associated disorder may vary according to a number of factors e.g. the particular gene(s) or genetic variation(s) in question; the total genetic environment;[299] and environmental factors. Other factors that might affect a person's likelihood of developing a particular disorder remain unknown.

Even if a genetic mutation is predictive or fully penetrant, the genetic test results cannot always, at least at this stage, predict the mildness or severity of the expression of the disorder in the particular person tested. Additionally, there are no guarantees as to the age of onset, or indeed if onset will occur at all, for any of these types of conditions.

### 3.6 Discussion and conclusion

The same limited body of evidence exists against which to judge the effects of genetic testing both of minors who can give their own consent, and those who cannot. However, two very different conclusions have been reached, because of the different consequences of testing each group.

Testing on a competent minor's request may be beneficial and may even enhance that minor's competence and autonomy. However, when testing of a child who cannot give informed consent is not clinically indicated, there is reason to suspect that psychological or social harms may arise. These harms could arise from early knowledge that one will inherit an untreatable disorder; because one has had no say in whether or not to be tested; because parents may treat their child differently to the detriment of the child; or because of a lack of ability to prevent parental dissemination of one's genetic information. Genetic testing for non-medical reasons, as a rule, should not be performed on minors who cannot give their own informed consent to testing because of the lack of evidence of the effects of genetic testing of minors; the fact that many adults choose not to discover their own genetic risk status; and the threats to a child's autonomy and confidentiality. Where there are no medical benefits to genetic testing, such testing should be delayed until the minor can give consent to the testing.

## ENDNOTES

1 Colin L. Soskolne, 'Beneficence,' *Gale Encyclopedia of Public Health*, The Gale Group Inc., Macmillan Reference, New York, 2002, accessed on 8 January 2007 from www.healthline.com/galecontent/beneficence?utm_term=beneficence&utm_medium=mw&utm_campaign=article.

2 See T.L. Beauchamp and J.F. Childress (eds) *Principles of Biomedical Ethics* Fifth Edition, Oxford University Press, New York, 2001 for discussion of the three major principles of bioethics: beneficence, non-maleficence and autonomy.

3 See discussion above in section on 'Professional Guidelines and Position Papers in Respect of Genetic Testing of Minors', and Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 1.1, p 16, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), for an overview of the arguments presented in the major United States, United Kingdom, Australasian and European statements on genetic testing and minors.

4 J.K. Mason and G.T. Laurie *Mason and McCall Smith's Law and Medical Ethics*, (7th ed.) Oxford University Press, 2006, p 219.

5 For example, M. Bloch and M.R. Hayden (1990) 'Opinion: Predictive Testing for Huntington Disease in Childhood: Challenges and Implications' *Am J Hum Genet* 46: 1–4, p 1 – 'Predictive testing for HD will serve as an important model for delivery of such a test for other late-onset incurable genetic disorders, and the guidelines developed in the former may be directly applicable to such programs.'

6 Cynthia B. Cohen 'Moving away from the Huntington's disease paradigm in the predictive genetic testing of children,' p 133–143, 134, in Angus Clarke (ed.) *The Genetic Testing of Children* BIOS Scientific Publishers Ltd, Oxford, 1998.

7 Susan Hogben and Paula Boddington (2005) 'Policy Recommendations for Carrier Testing and Predictive Testing in Childhood: A Distinction That Makes a Real Difference' *Journal of Genetic Counseling* 14, 4, August, 271–281, 277. Their special interest is the distinction drawn between carrier testing and predictive testing, and they criticise the role of the HD example in minimising carrier testing 'as "less serious" through implied contrast.'

8 Susan Hogben and Paula Boddington (2005) 'Policy Recommendations for Carrier Testing and Predictive Testing in Childhood: A Distinction That Makes a Real Difference' *Journal of Genetic Counseling* 14, 4, August, 271–281, 277.

9 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 1.1, p 16, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

10 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 96, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006).

11 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p iii, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006).

12 Julia Binedell (1998) 'Adolescent requests for predictive genetic testing,' pp123–132 in Angus Clarke (ed.) *The Genetic Testing of Children* BIOS Scientific Publishers Ltd, Oxford, 1998, cites findings from a European Community Collaborative HD Study (1993) that self-referral by adolescents does occur, although rarely. See Table 1, p 125. Additionally Duncan et al. surveyed 301 respondents from the United States, Canada, the United Kingdom, Australia and New Zealand who were professionally involved in predictive genetic testing as to their involvement and attitudes in relation to predictive genetic testing of pre-symptomatic children for untreatable conditions; and their views on the

existing guidelines on the predictive testing of children. Of the twenty-seven reported cases of testing of mature young people, in four cases (15 per cent) the young person's parents requested the testing, and in ten cases (37 per cent) the young person requested the testing. In the remaining thirteen cases (48 per cent) the request for testing came from both the young person and the parents. The survey undertaken by Duncan et al. provides the most recent evidence of professional practice in relation to genetic testing of minors. It is also one of the most geographically broad surveys. (Rony Duncan, Julian Savulescu, Lynn Gillam, Robert Williamson and Martin Delatycki (2005) 'An international survey of predictive genetic testing in children for adult onset conditions' *Genetics IN Medicine* Vol 7, No 6, 390–396.)

13 That is the age of majority in many countries, including the United Kingdom, but not in New Zealand where the age of majority is twenty (Age of Majority Act 1970). However, the Care of Children Act 2004 provides that guardianship ends at age eighteen and that is the age at which persons are legally entitled to do most things.

14 Sharpe et al. (1993) 'Letter to the Editor: Presymptomatic Testing for Huntington Disease: Is There a Duty to Test Those Under the Age of Eighteen Years?' *American Journal of Medical Genetics* 46: 250–253, 251.

15 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 85, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006).

16 Ellen Wright Clayton (1997) 'Genetic Testing in Children' *The Journal of Medicine and Philosophy* 22: 233–251, pp 240–241.

17 Angus Clarke and Frances Flinter 'The genetic testing of children: A clinical perspective' pp 164–176, 164–165 in Theresa Marteau and Martin Richards (eds.) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996. However, Bloch and Hayden have cautioned that predictive testing is not always an uncontroversial way to diagnose a disease for which one appears to be symptomatic. For example if a person presented with neurological or behavioural change and requested predictive testing for HD (perhaps because his parent has HD or is at risk of HD), the danger is 'that symptoms that are not due to HD may be wrongfully attributed to the early manifestations of the disorder.' M. Bloch and M.R. Hayden (1990) 'Opinion: Predictive testing for Huntington Disease in Childhood: Challenges and Implications' *Am J Hum Genet* 46: 1–4, p 2. Even if the person tested positive for the HD mutation the question of whether the present symptoms were due to HD could only be decided by a detailed clinical examination.

18 The focus is on predictive testing for *late* onset disorders in this section because it is likely that if a minor was at-risk for an *early* onset disorder she would be symptomatic by the time she was approaching competency if she had inherited the disorder.

19 There are varying accounts of whether the inherited mutation in the APC gene is 100 per cent penetrant: that is, whether every person with a copy of the mutated gene will develop FAP during their life. Some suggest it is only 90 per cent penetrant, while others speak as if it is 100 per cent penetrant. Rony Duncan states that 'Young people who are gene-positive for FAP are certain to develop cancer later in life if they remain untreated' (Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 5, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), referring to A.M. Codori, K.L. Zawacki, G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects' *Am J Med Genet* 2003;116A(2):117-28, E.W. Naylor and E.J. Gardner (1977) 'Penetrance and expressivity of the gene responsible for the Gardner syndrome' *Clin Genet* 1977;11:381-393, and S. Bulow (1987) 'Familial polyposis coli' *Dan Med Bull* 1987; 34:1-15.)

20 Centre for Genetics Education, *Genetics Fact Sheet – Bowel Cancer – Genetic Aspects,* accessed on 16 September 2006 from www.genetics.com.au/factsheet/46.htm.

21 Centre for Genetics Education, *Genetics Fact Sheet – Bowel Cancer – Genetic Aspects,* accessed on 16 September 2006 from www.genetics.com.au/factsheet/46.htm.

22 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 5, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006), referring to B.S. Elger and T.W. Harding (2000) 'Testing Adolescents for a Hereditary Breast Cancer Gene (BRCA1)' *Arch Pediatr Adolesc Med* 2000;154(2):113-119, 118, and Julian Savalescu (2001) 'Predictive genetic testing in children' *MJA* 175; 379–381, Box 2, accessed on 8 May 2006 from www.mja.com.au/public/issues/175 07 011001/savulescu/savulescu.html. Competency is discussed in greater detail below in the section on *Genetic Testing of Competent Minors.*

23 Allyn McConkie-Rosell and Gail A. Spiridigliozzi (2004) 'Family Matters: A Conceptual Framework for Genetic Testing in Children' *Journal of Genetic Counseling*, Vol. 13, No. 1, February 2004, p 13, citing BS Elger and TW Harding (2000) 'Testing Adolescents for a Hereditary Breast Cancer Gene (BRCA1)' *Arch Pediatr Adolesc Med* 2000;154(2):113-119, 118, and Angus Clarke and Frances Flinter 'The genetic testing of children: a clinical perspective' pp 164-176, 164-165 in Theresa Marteau and Martin Richards (Eds.) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996. Michie and Marteau also argue that 'research has long-since confirmed the common-sense belief that children need to exercise choice in order to acquire self-respect and a sense of responsibility' (Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 6, citing A.M. Clarke and A.D.B. Clarke (eds) *Early Experience: Myth and Evidence*, London, Open Book, 1976.

24 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf). See also Angus Clarke ('Genetic testing of children' in Norio Fujiki and Darryl R.J. Macer (eds) *Intractable Neurological Disorders, Human Genome Research and Society. Proceedings of the Third International Bioethics Seminar in Fukui, 19-21 November, 1993*, 218-221, accessed on 16 April 2006 from www.eubios.info/IND/INDAC.htm) who argues that granting control to the child could be helpful in enhancing her self-esteem, and in allowing her to come to terms with possibly unfavourable results. See also Stephen Robertson and Julian Savulescu (2001) 'Is There a Case in Favour of Genetic Testing of Young Children' *Bioethics* 15:1, 26, 49, who state that 'Testing could be viewed by some as a necessary step *towards* the attainment of autonomy.'

25 Julian Savalescu (2001) 'Predictive genetic testing in children' *MJA* 175; 379-381, Box 2, accessed from www.mja.com.au/public/issues/175 07 011001/svaulescu/savulescu.html 8 May 2006.

26 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

27 See Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf); Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 9; and S. Michie, M. Bobrow, and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: a study of emotional impact' *J Med Genet* 38(8):519-26. Minors who

know their HIV status have been reported to have higher self-esteem than children who are unaware of their status: Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 73, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), referring to Committee on Pediatric AIDS (1999) 'Disclosure of Illness Status to Children and Adolescents with HIV Infection' *American Academy of Pediatrics* 103(1):164–166.

28 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

29 Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 23.

30 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 36 (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006), J. Binedell, J.R. Soldan, J. Scourfield, and P.S. Harper (1996) 'Huntington's disease predictive testing: the case for an assessment approach to requests from adolescents.' *J Med Genet* 1996;33(11):912-8, 913, S. Michie, M. Bobrow, and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, and Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 6.

31 Julian Savalescu (2001) 'Predictive genetic testing in children' *MJA* 175; 379–381, Box 2, accessed www.mja.com.au/public/issues/175 07 011001/svaulescu/savulescu.html 8 May 2006, Stephen Robertson and Julian Savulescu (2001) 'Is There a Case in Favour of Genetic Testing of Young Children' *Bioethics* 15:1, 26, and Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 9.

32 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

33 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

34 Angus Clarke 'Genetic testing of children' in Norio Fujiki and Darryl R.J. Macer (eds) *Intractable Neurological Disorders, Human Genome Research and Society. Proceedings of the Third International Bioethics Seminar in Fukui, 19–21 November, 1993*, 218–221, accessed on 16 April 2006 from www.eubios.info/IND/INDAC.htm.

35 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

36 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), and S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: a study of emotional impact' *J Med Genet* 38(8):519–26.

37 Most of the following benefits of mutation negative results are incorporated from Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006).

38 Angus Clarke 'Genetic testing of children' in Norio Fujiki and Darryl R.J. Macer (eds) *Intractable Neurological Disorders, Human Genome Research and Society. Proceedings of the Third International Bioethics Seminar in Fukui, 19-21 November, 1993*, 218–221, accessed on 16 April 2006 from www.eubios.info/IND/INDAC.htm and Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 9.

39 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), and Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 9.

40 Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, p 5. A woman who gave her personal experience of being at-risk for the HD mutation said 'In practice, being "at risk" meant that I was treated in the same way as if I definitely had the gene.'

41 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.2, p 218, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

42 Barbara A. Bernhardt, Ellen S. Tambor, Gertrude Fraser, Lawrence Wissow and Gail Geller (2003) 'Parents' and Children's Attitudes Toward the Enrollment of Minors in Genetic Susceptibility Research: Implications for Informed Consent' *American Journal of Medical Genetics* 116A: 315-323, 321. One woman recalled being caught 'off-guard' by her own breast cancer diagnosis and having felt rushed into making uninformed decisions. 'A positive genetic test result would allow time for research and adjustment: "more informed decision-making. Time to make good decisions and to get information gathered. … when I got my diagnosis, I didn't feel like I had the time … I didn't take time to do the research because I wasn't emotionally ready to do that."'

43 Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, pp 6, 13.

44 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

45 Fanos refers to a study of seventy-five adult siblings of persons affected by cystic fibrosis. They found that over a third of the siblings had significant misunderstandings regarding their genetic risks: e.g. a thirty-three year old had been told he was the 'strongest carrier' in his family, and so decided not to have children. (Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, p 23, referring to J.H. Fanos (1987) 'Developmental Consequences for Adulthood of Early Sibling Loss' *Dissertations Abstracts International,* 48-08B, number 8723871, and J.H. Fanos and B.G. Nickerson 'Long-term effects of sibling death during adolescence' in D Balk (ed.) (1991) 'Special Issue, Death and Adolescent Bereavement' *J Adol Res* 6:70–82.)

46 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

47 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 27, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), on 17 September 2006.

48 Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 26.

49 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

50 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 26, and D.C. Wertz, J.H. Fanos, and P.R. Reilly (1994) 'Genetic testing for children and adolescents. Who decides?' *JAMA* 1994;272(11):875-81, 876.

51 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

52 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

53 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 26, and D.C. Wertz, J.H. Fanos and P.R. Reilly (1994) 'Genetic testing for children and adolescents. Who decides?' *JAMA* 1994;272(11):875-81, 876.

54 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

55 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

56 D.C. Wertz, J.H. Fanos and P.R. Reilly (1994) 'Genetic testing for children and adolescents. Who decides?' *JAMA* 1994;272(11):875-81, 876.

57 Allyn McConkie-Rosell and Gail A. Spiridigliozzi (2004) 'Family Matters: A Conceptual Framework for Genetic Testing in Children' *Journal of Genetic Counseling*, Vol. 13, No. 1, February 2004, p 12.

58 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 23, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006), and Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 26.

59 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), for the four points below.

60 Human Genetics Society of Australasia Policy *Predictive Testing in Children and Adolescents*, Version 2, April 2005, accessed on 8 May 2006 from http://hgsa.com.au/images/UserFiles/Attachments/Predictivetesting(General)APRIL2005.pdf.

61 D.C. Wertz, J.H. Fanos and P.R. Reilly (1994) 'Genetic testing for children and adolescents. Who decides?' *JAMA* 1994;272(11):875-81, 876.

62 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 23, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006), and Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 27.

63 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

64 Human Genetics Society of Australasia Policy *Predictive Testing in Children and Adolescents*, Version 2, April 2005, accessed on 8 May 2006 from http://hgsa.com.au/images/UserFiles/Attachments/Predictivetesting(General)APRIL2005.pdf, Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, Table 7.1, p 210, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006), and Allyn McConkie-Rosell and Gail A. Spiridigliozzi (2004) 'Family Matters: A Conceptual Framework for Genetic Testing in Children' *Journal of Genetic Counseling*, Vol. 13, No. 1, February 2004, p 12.

65 Human Genetics Society of Australasia Policy *Predictive Testing in Children and Adolescents*, Version 2, April 2005, accessed on 8 May 2006 from http://hgsa.com.au/images/UserFiles/Attachments/Predictivetesting(General)APRIL2005.pdf, D.C. Wertz, J.H. Fanos, and P.R. Reilly (1994) 'Genetic testing for children and adolescents. Who decides?' *JAMA* 1994;272(11):875-81, 876, Allyn McConkie-Rosell and Gail A. Spiridigliozzi (2004) 'Family Matters: A Conceptual Framework for Genetic Testing in Children' *Journal of Genetic Counseling*, Vol. 13, No. 1, February 2004, p 12. See also Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 26 : 'Since the adolescent has begun to relinquish parental attachments and does not yet have adult primary attachments, it is a time of great vulnerability and desire for peer approval.'

66 Allyn McConkie-Rosell and Gail A. Spiridigliozzi (2004) 'Family Matters: A Conceptual Framework for Genetic Testing in Children' *Journal of Genetic Counseling*, Vol. 13, No. 1, February 2004, p 13, D.C. Wertz, J.H. Fanos and P.R. Reilly (1994) 'Genetic testing for children and adolescents. Who decides?' *JAMA* 1994; 272(11):875-81, 876, and Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 26.

67 Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 27.

68 Many of these examples are taken from Stephen Robertson and Julian Savulescu (2001) 'Is there a Case in Favour of Predictive Genetic Testing in Young Children?' *Bioethics* Vol. 15, No. 1, 2001, p 34. Also see Human Genetics Society of Australasia Policy *Predictive Testing in Children and Adolescents*, Version 2, April 2005, accessed on 8 May 2006 from http://hgsa.com.au/images/UserFiles/Attachments/Predictivetesting(General)APRIL2005.pdf.

69 We refer to a lack of pre-symptomatic medical benefits specifically because, in line with the points raised above, there are benefits to symptomatic testing: to identify the disorder and disqualify other disorders, resulting in a more focussed response (regardless of whether or not that response includes clinically beneficial outcomes).

70 Centre for Genetics Education, *Genetics Fact Sheet – Huntington Disease,* accessed on 16/09/06 from www.genetics.com.au/factsheet/37.htm.

71 Clinical Genetics Society (UK) (1994) 'The Genetic Testing of Children, Report of a Working Party of the Clinical Genetics Society, Chaired by Dr Angus Clarke' J *Med Genet* 1994; 31; 785-797, 791.

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75 Information accessed on 8 January 2007 from www.southerncross.co.nz/index.cfm?316B8738-F964-4D19-BBA1-D6301B50B611.

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77 Centre for Genetics Education, *Genetics Fact Sheet – Breast and Ovarian Cancer – Genetic Aspects*, accessed on 16 September 2006 from www.genetics.com.au/factsheet/36.htm.

78 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 31, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006), referring to B.S. Elger and T.W. Harding (2000) 'Testing Adolescents for a Hereditary Breast Cancer Gene (BRCA 1)' *Arch Pediatr Adolesc Med* 154(2), 113-119, p 117.

79 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 25, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006), quoting J. Holland (1997) 'Should Parents be Permitted to Authorize Genetic Testing For Their Children?' *Fam Law Quart* 31(2), 321-353, at 338.

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'Breast and ovarian cancer incidence in BRCA1-mutation carriers' *Am J Hum Genet* 56: 265-271; and D. Ford, D.F. Easton, D.T. Bishop, S.A. Narod, and D.E. Goldgar (1994) Breast Cancer Linkage Consortium 'Risks of cancer on BRCA1-mutation carriers' *Lancet* 343: 692-695. Asaff Harel, Diane Abuelo (MD) and Alessandra Kazurav (MD) (2003) 'Adolescents and Genetic Testing: What Do They Think About It?' *Journal of Adolescent Health* 33: 489-494 referring to R. Wooster, S.L. Neuhausen and J. Mangion et al. (1994) 'Localization of a breast cancer susceptibility gene, BRCA2, to chromosome 13q-12-13' *Science* 265: 2088-90.

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97 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 249 (accessed 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

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99 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005 (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006), and Human Genetics Society of Australasia Policy *Predictive Testing in Children and Adolescents*, Version 2, April 2005, accessed on 8 May 2006 from http://hgsa.com.au/images/UserFiles/Attachments/Predictivetesting(General)APRIL2005.pdf.

100 In terms of how minors adapt to illness, there is some evidence that minors who learn of their compromised health earlier rather than later adapt better. Having hypothesised that childhood cancer survivors who had learned of their diagnosis at an early stage would tend to be better adjusted at follow-up than those who learned they had cancer long after the diagnosis was made, Slavin et al. studied 116 survivors on average twelve years after diagnosis. They found that good psychological adjustment was associated with patients' early knowledge of the diagnosis, and that many of the survivors, their parents and their siblings felt that a cancer diagnosis should be shared with a child early on. Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 6, referring to L.A. Slavin, M.D. O'Malley, G.P. Koocher, and D.J. Foster (1982) 'Communication of the cancer diagnosis to pediatric patients: Impact on long-term adjustment' *American Journal of Psychiatry*, 139, 179-183.

101 See Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 49-51,

(accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), referring to M. Shepherd, A.T. Hattersley, and A.C. Sparkes (2000) 'Predictive genetic testing in diabetes: A case study of multiple perspectives' *Qual Health Res* 10(2):242-59, M. Shepherd, I. Ellis, A.M. Ahmad et al. (2001) 'Predictive genetic testing in maturity-onset diabetes of the young (MODY)' *Diabetic Medicine* 18(5):417-21, and S. Michie, V. McDonald, M. Bobrow, C. McKeown, and T. Marteau (1996) 'Parents' Responses to Predictive Genetic Testing in Their Children – Report of a Single Case Study' *J Med Genet* 33(4):313-318.

102 Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, referring to S.B. Johnson 'The psychological impact for risk screening of chronic and life-threatening childhood illnesses' in J.H. Johnson and S.B. Johnson (eds) *Advances in Child Health Psychology*, University of Florida Press, 1991.

103 Information accessed on 8 January 2007 from http://en.wikipedia.org/wiki/Hyperlipidemia.

104 The researchers used the standard Child Behaviour Checklist Tool (CBCL) for measuring behaviour problems. 'The CBCL is a 118-item checklist that measures behaviour problems and competencies. Problems include somatic complaints, attention difficulties, and delinquent behaviour; competencies include recreational activities, social behaviour, and school functioning.' A.M. Codori, K.L. Zawacki, G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: long-term psychological effects' *Am J Med Genet* 2003;116A(2):117-28, 119.

105 Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, referring to E. Rosenberg, D.L. Lamping, L. Joseph, S. Blaichman, B. Pless, and M. Lambert (1992) 'Cholesterol Screening of High-Risk Children: Psychological and Behavioural Effects' Unpublished Report to the Fonds de la Recherche en Sante du Quebec (FRSQ).

106 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt, F.M. Giardiello (1996) 'Genetic testing for cancer in children. Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11), 1131-38. Parents gave informed consent for their children to participate in the study, and all children assented.

107 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt, F.M. Giardiello (1996) 'Genetic testing for cancer in children. Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11), 1131-38, 1136.

108 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt, F.M. Giardiello (1996) 'Genetic testing for cancer in children. Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11), 1131-38, 1135. See note 105 for a description of the CBCL used to measure behaviour. To address the possibility that mutation-positive children were not reporting clinically significant increases in distress because they did not understand the implications of their diagnosis, correlations between age and distress were examined: there was no significant association (1136).

109 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt, F.M. Giardiello (1996) 'Genetic testing for cancer in children. Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11), 1131-38, 1136-37. It was difficult to draw a conclusion on those particular results, although the authors considered identification and mimicking, and attachment theory as potentially relevant. The authors acknowledged that it was 'premature to conclude that long-term follow-up will be equally favourable'. They also noted that the psychological effect of testing may have been modified by the provision of psychological support to those children most likely to have difficulty after testing (p 1131).

110 S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, 520.

111 Mean scores for anxiety among adults who tested negative for the FAP mutation were within the normal range. The only difference between the anxiety and depression scores amongst the adults and children was that amongst those with positive test results, the adults were more anxious than the children, and fewer children than adults had anxiety scores in the clinical range, S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, 521-523.

112 S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, 523.

113 S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, 524-525. Other possibilities for the difference between adult and child levels of anxiety include: adults have been found to have a greater bias towards being unrealistically optimistic about their health risks than adolescents, perhaps resulting in raised anxiety on receipt of a positive test result, having had a greater expectation of a negative result. Additionally, children may receive more support from health professionals and family members, both before and after testing. However, the authors also note that two aspects of their research method may have added to the difference: clinical geneticists may have screened out those children who seemed too distressed or vulnerable to participate; and the small sample size might mean that the study was underpowered (S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26).

114 S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, 524.

115 S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, 526.

116 A.M. Codori, K.L. Zawacki and G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects.' *Am J Med Genet* 116A(2), 117-28. Data was collected before testing, at 3, 12 and 23-55 months after disclosure of the test results. The mean length of time of follow-up was 38 months (see p 117). Length of follow-up was unrelated to the outcome variables (p 120).

117 A.M. Codori, K.L. Zawacki and G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects.' *Am J Med Genet* 116A(2), 117-28, 117.

118 'Our long-term follow-up data suggest that testing is not associated with clinically significant increases in mean depression or anxiety symptoms, increases in mean behaviour problems …. Nonetheless, it would be clinically irresponsible to ignore the increases in depression and anxiety among children with positive siblings. … our data suggest that the mixed families and those where more than one child tests positive are in need of support long after disclosure of the test results.' A.M. Codori, K.L. Zawacki and G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: long-term psychological effects.' *Am J Med Genet* 116A(2), 117-28, 117.

119 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 53, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf, on 17 September 2006). We refer frequently to the extensive research recently undertaken by Rony Duncan into the purported harms of predictive testing in young people: in particular, Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006; Rony Duncan, Julian Savulescu, Lynn Gillam, Robert Williamson, and Martin Delatycki (2005) 'An international survey of predictive genetic testing in children for adult onset conditions' *Genetics IN Medicine* Vol. 7, No. 6, 390-396. In her Ph.D thesis Duncan offers a meta-analysis of the professional guidelines, position statements, and academic literature on genetic testing and young people (and also predictive testing for HD, more generally). She also presents her own empirical research on current professional practice around genetic testing of minors, and the effects of predictive testing for FAP and HD for young people.

120 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 53-54, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf). Duncan postulates six reasons why empirical

research has thus far failed to adequately address the potentially beneficial outcomes of genetic testing: 1) researchers may believe that finding evidence of harmful effects (or a lack of evidence of harmful effects) will be more powerful than finding evidence of beneficial effects (or a lack of evidence of beneficial effects); 2) researchers have preconceived ideas about the types of impacts that predictive genetic testing will have; 3) researchers are simply choosing to test the hypotheses that are most prominent in current debates; 4) researchers are choosing to use the most standardised and widely accepted instruments for assessing psychological traits; 5) researchers are measuring the types of outcomes that have been most used in research concerning testing of adults; 6) researchers believe the outcomes they have chosen are able to demonstrate both harm and benefit.

121 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 56 (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006 – predictive testing for HD via linkage analysis has been available since 1986, and direct gene testing since 1993. Duncan refers to M.B. Delatycki and R. Tassicker (2001) 'Adult Onset Neurological Disorders: Predictive Genetic Testing' *Aus Fam Physic* 30(10), and E. Almqvist, R. Brinkman, S. Wiggins and M. Hayden (2003) 'Psychological consequences and predictors of adverse events in the first 5 years after predictive testing for Huntington's disease' *Clin Genet* 64(4):300-309.

122 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 57 (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006), referring to R. Timman, R. Roos, A. Maat-Kievit and A. Tibben (2004) 'Adverse Effects of Predictive Testing for Huntington Disease Underestimated: Long-Term Effects 7-10 Years After the Test.' *Health Psychol* 2004;23(2):189-197.

123 See Duncan's literature review at pp 56-61: Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

124 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 63, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006).

125 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 57, (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006).

126 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 57-58 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

127 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 64 (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006.

128 Rony Duncan, Julian Savulescu, Lynn Gillam, Robert Williamson, and Martin Delatycki (2005) 'An international survey of predictive genetic testing in children for adult onset conditions' *Genetics IN Medicine* Vol. 7, No. 6, 390-396.

129 Information accessed on 8 January 2007 from www.mda.org.au/specific/mdamyt.html.

130 Information accessed on 8 January 2007 from http://en.wikipedia.org/wiki/Spinocerebellar_ataxia.

131 Rony Duncan, Julian Savulescu, Lynn Gillam, Robert Williamson, and Martin Delatycki (2005) 'An international survey of predictive genetic testing in children for adult onset conditions' *Genetics IN Medicine* Vol. 7, No. 6, 390-396, 394, and Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 139-140 (accessed on 17 September from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

132 Rony Duncan, Julian Savulescu, Lynn Gillam, Robert Williamson and Martin Delatycki (2005) 'An international survey of predictive genetic testing in children for adult onset conditions' *Genetics IN Medicine* Vol. 7, No. 6, 390-396, 394.

133 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 113-117 (accessed on 17 September from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.). She had initially hoped to interview persons who had undergone predictive testing under the age of eighteen for non-medical reasons, but could find very few respondents who fitted those criteria (perhaps because of the HGA guidelines and popular discourse against offering such testing). She therefore widened her criteria to include minors who had undergone testing for medical reasons (e.g. for FAP) and also persons who had undergone predictive testing for HD who had been over eighteen but still a young person on some definitions at the time of testing (people under the age of twenty-five years).

134 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 159 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

135 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 159 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

136 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 159 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

137 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 211 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

138 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 216 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

139 For example, see Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 161 and 192, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

140 Dorothy C. Wertz (2000) 'Genetic Testing of Children: British Medical Association – EUROSCREEN Conference, London, United Kingdom, 13–15 June, 1996' in *GeneLetter*, 1 May, accessed 10 May 2006 from www.genesage.com/professionals/geneletter/archives/testingchildren.html, and also Dorothy C Wertz (2000) 'Testing children: Pros and Cons' in *GeneLetter*, 1 May, accessed 10 May 2006 from www.genesage.com/professionals/geneletter/05-01-00/features/children.html.

141 For example, see Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005,

pp 156 and 187, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

142 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 186 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

143 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 185 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

144 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 192 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

145 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 189 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

146 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 188 and 215 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

147 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 213 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

148 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 170 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.) Also, 'If I had of had the test done now, it would have had a bigger impact on me than what it would have six years ago because I think if you are younger you don't really sort of have the same sort of knowledge that you have when you're older and things can have a bigger impact on you' (p 216). See also, Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, p 23: 'I first heard about Huntington's disease … I was 15 years old. … From the diary which I kept at the time, it would seem the initial impact this information had on me was relatively slight, with other aspects of my life continuing to dominate my thoughts.'

149 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 178 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

150 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 197 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

151 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 178 and 195 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

152 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 178 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

153 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 179 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

154 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 196 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

155 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 178 and 244 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

156 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 192 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

157 For example, 'Mum can breathe too,' and 'They were happy, they were happy,' Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 215 (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf) on 17 September 2006).

158 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 190 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

159 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 179 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

160 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 188 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

161 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 206 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

162 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 171 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

163 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 180 and 208 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

164 Interestingly, this comment came from a young man who tested positive for the FAP mutation, a procedure which has medical benefits: what he did not know, if he had not been tested, may well have hurt him, particularly if he did not keep up appropriate surveillance.

165 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 240 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

166 'I would have loved to keep catching up with her and trying to work out, ok, this is what you do from here, yeah, but I've been left to go off and work it out for myself and I find that really hard, really frustrating, cause what to do?' Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 208 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.) '… I was meant to be contacted in regards of going down again afterwards … but that never happened … which probably would have been good…um, to just say yeah I've been pretty down, like, or I've been pretty flat over the, for a month kind of thing and, just to be told yes, that's normal would have been nice…' (p 240).

167 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 207 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

168 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 190 and 206 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

169 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 207 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

170 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 174 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

171 'Telling people is just really really hard, you don't know how they're going to react, like when I told my grandfather … by the end … he was almost crying.' Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 198 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.) 'Everyone at work … it's just phenomenal, oh, yap yap yap yap yap, such and such is dying from this, dying from that … they all just create numerous stories.' (p 205). 'I've been let down by a couple of people who, you know, who are… thoughtless, just the complete thoughtlessness and selfishness.' (p 206).

172 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 188 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

173 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 206 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

174 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 180 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

175 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 180 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

176 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 207 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

177 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 180 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

178 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 183 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

179 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 186 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

180 For example, 'If I didn't have it, I'd be much happier … less worried about things … cause I'm worried about all my health … like, a tumour or something …' Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 183 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

181 For example, 'I was crying and oh I was so upset…' Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 183 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

182 For example, 'I actually got a bit angry … shaky, ah, ok, very down, depressed' Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 183 (accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006.)

183 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 185 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

184 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 184 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

185 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 205 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

186 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 205 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

187 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 198 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

188 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 205 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

189 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 188 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

190 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 198 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

191 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 248 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

192 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 163 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

193 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 198 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

194 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 237 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

195 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 223-227 (accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.)

196 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt and F.M. Giardiello (1996) 'Genetic testing for cancer in children: Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11):1131-8 and A.M. Codori, K.L. Zawacki and G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects.' *Am J Med Genet* 116A(2), 117-28.

197 For example, 'I sort of wasted my year 12 away so badly … I'm really upset for that … but I honestly, honestly thought I was going to get the gene, honestly.' Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 230, accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf.) 'Just to stop the devastation, just to gear myself up to deal with it … my driver's licence will be slowly cut down and

I'm not allowed to get life insurance and … I've wanted to be a lawyer some of my life and go and join the army but no way would I get in and … I'm going to get kicked back over and over again as the years go on so gear yourself up, you know, you might as well start preparing now … cause if the worst doesn't happen, only good can come from there I thought.' (p 230). 'I knew I would have it … I know it's got nothing to do with it, but everyone's always said … that I was exactly like my father, I look like him, I talk like him, I'm outgoing like he is … so I knew … I know there's a 50–50 chance, but I knew I'd have it … I wouldn't say I was shocked, because … I expected it, I really did.' (p 230).

198 Quote from a young adult woman in Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, p 16.

199 Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1: 3-14, p 12.

200 We use the word 'historical' loosely to indicate that most of the papers referred to were written more than fifteen years ago, which makes them somewhat historic given the rapid increase in knowledge about human genetics over the last fifteen years.

201 Angus Clarke 'Genetic testing of children' in Norio Fujiki and Darryl R.J. Macer (eds) *Intractable Neurological Disorders, Human Genome Research and Society. Proceedings of the Third International Bioethics Seminar in Fukui, 19-21 November, 1993*, 218-221, accessed on 16 April 2006 from www.eubios.info/IND/INDAC.htm.

202 Angus Clarke 'Genetic testing of children' in Norio Fujiki and Darryl R.J. Macer (eds) *Intractable Neurological Disorders, Human Genome Research and Society. Proceedings of the Third International Bioethics Seminar in Fukui, 19-21 November, 1993*, 218-221, accessed on 16 April 2006 from www.eubios.info/IND/INDAC.htm.

203 G. Stamatoyannopoulus (1974) 'Problems of screening and counseling in the haemoglobinopathies' in A.G. Motulsky and F.J.B. Ebling (eds) *Birth Defects: Proceedings of the Fourth International Conference* Amsterdam, Excerpta Medica.

204 T.M. Marteau et al. (1992) 'Effects of genetic screening on perceptions of health: a pilot study' *J Med Genet* 29, 24-26.

205 S. Zeesman et al. (1984) 'A private view of heterozygosity: Eight-year follow-up study on carriers of the Tay-Sachs gene detected by high school screening in Montreal.' *Am J Med Genet* 18: 769-778.

206 S. Loader et al. (1991) 'Prenatal hemoglobinopathy screening IVF follow-up of women at risk for a child with a clinically significant hemoglobinopathy.' *Am J Hum Genet* 49: 1292-1299.

207 J.E. Bowman (1991) 'Invited editorial: Prenatal screening for haemoglobinopathies.' *Am J Hum Genet* 48: 433-438, and Kenen and Schmidt (1978) 'Stigmatisation of carrier status: Social implications of heterozygote screening programmes.' *Am J Pub Health* 68: 1116-1120. '(T)here are sufficient historical data to substantiate the concern when African Americans were compelled in some American states to undergo testing for sickle cell diseases in the 1970s, and were subsequently discriminated against on the basis of carrier status.': Canadian Paediatric Society, Position Statement (B 2003 – 01) 'Guidelines for genetic testing of healthy children,' in *Paediatrics and Child Health* Vol. 8 No. 1 January 2003, 42-45, 43, referring to President's Commission for the study of ethical problems in medicine and biomedical and behavioural research; *Screening and Counseling for Genetic Conditions: A report on the ethical and legal implications of genetic screening, counselling and education programs.* Washington: President's Commission, 1983.

208 R.C. Trembath et al. (1991) 'Carrier testing for CFTR gene mutations in relatives identified through a cystic fibrosis prenatal testing service. Response rates and determinants of uptake.' *Clinical Genetics Society,* Belfast.

209 Angus Clarke and Frances Flinter 'The genetic testing of children: a clinical perspective' in Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, p 171, citing research by F.A. Flinter, A. Silver, C.G. Mathew and M. Bobrow (1992) 'Population screening for cystic fibrosis' *Lancet*, 339, 1539-40.

210 Dorothy C. Wertz (2000) 'Testing children: Pros and Cons' in *GeneLetter*, 1 May, accessed 10 May 2006 from www.genesage.com/professionals/geneletter/05-01-00/features/children.html.

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215 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt and F.M. Giardiello (1996) 'Genetic Testing for Cancer in Children' *Arch Pediatr Adolesc Med.* 1996 Nov;150(11):1131-8, p 1132, A.M. Codori, K.L. Zawacki, G.M. Petersen, D.L. Miglioretti, J.A. Bacon, J.D. Trimbath, S.V. Booker, K. Picarello and F.M. Giardiello (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects' *Am J Med Genet* A. 2003 Jan 15;116(2):117-28, and S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 2001;38:519-526 (August). Both parental informed consent and the child's assent were required for enrolment in Codori et al.'s studies. In Michie et al.'s study participants were asked to complete a consent form, and parental consent was required for participants aged under sixteen (in one centre, under eighteen).

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218 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 154, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

219 Rony E. Duncan, Julian Savulescu, Lynn Gillam, Robert Williamson and Martin B. Delatycki (2005) 'An international survey of predictive genetic testing in children for adult onset conditions' *Genetics IN Medicine* 7: 6: 390-396, p 395.

220 Rony E. Duncan, Julian Savulescu, Lynn Gillam, Robert Williamson and Martin B. Delatycki (2005) 'An international survey of predictive genetic testing in children for adult onset conditions' *Genetics IN Medicine* 7: 6: 390-396, p 395.

221 See a copy of the survey at www.mcri.edu.au/pages/Rony/Questionnaire2.asp?n.
222 Ellen Wright Clayton (1997) 'Genetic Testing in Children' *The Journal of Medicine and Philosophy* 22: 233-251, p 244.
223 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 262, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).
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225 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 262, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).
226 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 166, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).
227 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 260-261, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).
228 However, people who do not carry the genetic variation tested for are still at risk of developing a multifactorial disorder, such as cancer. For example, see Centre for Genetics Education, *Genetics Fact Sheet – Genetics and Cancer a*ccessed on 16 April 2007 from www.genetics.com.au/factsheet/44.htm: 'It is important to remember that if someone does not carry a mutation in a "cancer protection" gene that predisposes them to develop cancer they still have the same chance of developing cancer as anyone else in the community.'
229 Susan Hogben and Paula Boddington, Paula (2005) 'Policy Recommendations for Carrier Testing and Predictive Testing in Childhood: A Distinction That Makes a Real Difference' *Journal of Genetic Counseling* 14, 4, August, 271-281.
230 This research focuses on testing individual minors who are 'at risk' for a heritable genetic mutation, and not the effects of carrier screening programmes, which is why research into the outcomes of such programmes is not discussed at any great length.
231 K. Barlow-Stewart, L. Burnett, A. Proos, V. Howell, F. Huq, R. Lazarus and H. Alzenberg (2003) 'A genetic screening programme for Tay-Sachs disease and cystic fibrosis for Australian Jewish high school students' *J. Med. Genet* 40, 45. Despite efforts being made to avoid any element of coercion, including emphasising the voluntary nature of participation in the education session and in the written material, and the use of one-on-one consent interviews, 5 per cent of students who participated in the Tay-Sachs disease carrier screening programme indicated that peer pressure had influenced their testing choice.
232 Australian Law Reform Commission *Essentially Yours: The Protection of Human Genetic Information in Australia* accessed from www.austlii.edu.au/other/alrc/publications/reports/96/ 23 May 2006, para 24.42.
233 Australian Law Reform Commission *Essentially Yours: The Protection of Human Genetic Information in Australia* accessed from www.austlii.edu.au/other/alrc/publications/reports/96/ 23 May 2006, para 24.53.
234 In the limited studies that we have looked at it is usual for both parental consent and minor consent or assent to be sought before carrier screening takes place when the minors involved are under

the age of sixteen. This is unsurprising given the context of the screening taking place within the education system: there are professional liability issues for the health professionals and potentially also the education professionals, and time and efficacy difficulties involved in assessing the competency of each minor under the age of sixteen individually in order to see whether he or she can give informed consent on his or her own behalf.

235 K. Barlow-Stewart, L. Burnett, A. Proos, V. Howell, F. Huq, R. Lazarus and H. Alzenberg (2003) 'A genetic screening programme for Tay-Sachs disease and cystic fibrosis for Australian Jewish high school students' *J. Med. Genet* 40, 45.

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237 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 190, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

238 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 64, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

239 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 90, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

240 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 82, accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006).

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245 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 198, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

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250 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 185, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

251 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 166, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

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255 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 211, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

256 Susan Michie and Theresa M. Marteau (1996) 'Predictive genetic testing in children: The need for psychological research' *British Journal of Health Psychology* 1, 3-14, 6, citing A.M. Clarke and A.D.B. Clarke (eds) (1976) *Early Experience: Myth and Evidence*, London, Open Book.

257 Stephen Robertson and Julian Savulescu (2001) 'Is There a Case in Favour of Genetic Testing of Young Children' *Bioethics* 15:1, 26, 42.

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259 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt and F.M. Giardiello (1996) 'Genetic testing for cancer in children: Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11):1131-8, and A.M. Codori, K.L. Zawacki and G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects.' *Am J Med Genet* 116A(2), 117-28.

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261 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 216, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf). See also Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, p 23: 'I first heard about Huntington's disease … I was 15 years old. … From the diary which I kept at the time, it would seem the initial impact this information had on me was relatively slight, with other aspects of my life continuing to dominate my thoughts.'

262 S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26, 523.

263 Barbara A. Bernhardt, Ellen S. Tambor, Gertrude Fraser, Lawrence Wissow and Gail Geller (2003) 'Parents' and Children's Attitudes Toward the Enrollment of Minors in Genetic Susceptibility Research: Implications for Informed Consent' *American Journal of Medical Genetics* 116A: 315-323, 321.

264 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 31, accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006), referring to B.S. Elger and T.W. Harding (2000) 'Testing Adolescents for a Hereditary Breast Cancer Gene (BRCA 1)' *Arch Pediatr Adolesc Med* 154(2), 113-119, p 117.

265 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006): 'The community and the friendship … was so solid through the process' (p 215); '…it kind of brought us closer because my best friend, she has diabetes … I've got something and she's got something … we can talk to each other about it and at school that really helps' (p 188).

266 'Telling people is just really really hard, you don't know how they're going to react'; 'I've been let down by a couple of people who, you know, who are … thoughtless, just the complete thoughtlessness and selfishness'.

267 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 58-61, 63, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

268 Some might argue that: 'A substantial proportion of the tested children (often 33% or 50%) … will be reassured by favourable results from the genetic testing.' (Angus Clarke and Frances Flinter 'The genetic testing of children: A clinical perspective' in Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, p 168.) To some degree that argument uses statistics to argue for genetic testing of minors on the basis of a utilitarian calculation. Of course, as Clarke and Flinter point out, many would respond that 'the reassurance to (perhaps) half of the tested children is bought at too high a price: the unfavourable results given to the other children. (Angus Clarke and Frances Flinter 'The genetic testing of children: A clinical perspective' in Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and psychological implications of the new human genetics* Cambridge University Press, United Kingdom, 1996, p 168.) The United Kingdom Clinical

Genetics Society Report also notes that while 50 per cent of 'possible' carrier children would be shown not to be carriers if tested, and that life might be simplified for them, 'the possible benefits experienced by these children must be balanced against the possible harm done by identifying carriers in childhood.' (Clinical Genetics Society (UK) (1994) 'The Genetic Testing of Children, Report of a Working Party of the Clinical Genetics Society, Chaired by Dr Angus Clarke' J *Med Genet* 1994; 31; 785-797, 792.) The focus of this report is primarily autonomy and rights based: arguments offered in favour of recognising the right of competent minors to consent to or to refuse genetic testing do not rely upon a utilitarian framework for the greatest benefits for the greatest number. The decision is for each competent minor to make for him or herself as an autonomous individual. However, it is worth recalling that the odds are not stacked in favour of a mutation positive result, as often appears to be an underlying assumption in the discourse.

269 For example, see Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 231, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

270 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf on 17 September 2006): 'I mucked up a lot I think, cause I just thought I was going to get this illness,' (p 224); 'I got into drugs, I got onto heroin … it was a crutch for me,' (p 224); 'I didn't do well at school … just misbehaved really … I don't think I would have done it anyway,' (p 224) 'I used to drink a lot and get locked up and that … and I had depression and stuff … I left school at 15 … I just sort of kept to myself a lot, in me room and stuff … smoking and stuff …' (p 225).

271 Joanna H. Fanos (1997) 'Developmental Tasks of Childhood and Adolescence: Implications for Genetic Testing' *American Journal of Medical Genetics* 71:22-28, 24.

272 Sharpe et al. (1993) *Letter to the Editor*: ' Presymptomatic Testing for Huntington Disease: Is There a Duty to Test Those Under the Age of Eighteen Years?' *American Journal of Medical Genetics* 46:250-253, 250-251.

273 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 63, accessed from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), on 17 September 2006).

274 Sharpe et al. (1993) *Letter to the Editor*: 'Presymptomatic Testing for Huntington Disease: Is There a Duty to Test Those Under the Age of Eighteen Years?' *American Journal of Medical Genetics* 46:250-253, 251.

275 Pascal Borry, Jean-Pierre Fryns, Paul Schotsmans and Kris Dierickx (2006) 'Carrier testing in minors: A systematic review of guidelines and position papers' *European Journal of Human Genetics* 14, 133-138, 137.

276 Clinical Genetics Society (UK) (1994) **'**The Genetic Testing of Children, Report of a Working Party of the Clinical Genetics Society, Chaired by Dr Angus Clarke' *J Med Genet* 1994; 31; 785-797, 791.

277 Information accessed on 8 January 2007 from www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View..ShowSection&rid=gnd.section.143.

278 Information accessed on 8 January 2007 from www.retinoblastoma.net/whatisrb.html.

279 Information accessed on 8 January 2007 from www.ctf.org/index.php?option=com_content&task=view&id=29&Itemid=288.

280 Marc Tischkowitz and Elisabeth Rosser (2004) 'Inherited cancer in children: Practical/ethical problems and challenges' *European Journal of Cancer* 40, 2459-2470, p 2468.

281 *GIG Response to the Clinical Genetics Society Report: The Genetic Testing of Children* (1995), accessed 13 February 2007 from www.gig.org.uk/docs/gig_testingchildren.pdf.

282 Most of these examples are taken from Stephen Robertson and Julian Savulescu (2001) 'Is There a Case in Favour of Genetic Testing of Young Children' *Bioethics* 15:1, 26, 34.

283 For example, see Ellen Wright Clayton (1997) 'Genetic Testing in Children' *The Journal of Medicine and Philosophy* 22: 233-251, and Stephen Robertson and Julian Savulescu (2001) 'Is There a Case in Favour of Genetic Testing of Young Children' *Bioethics* 15:1, 26.

284 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 34-35, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf), referring to L.F. Ross and M.R. Moon (2000) 'Ethical issues in genetic testing of children.' *Arch Pediatr Adolesc Med* 2000;154(9):873-879.

285 Ellen Wright Clayton (1998) 'Genetic Testing in Children: The Law's Role in Addressing the Challenges Posed by New Technology (Commentary)' *Children's Legal Rights Journal* Vol. 18, No. 2, 39-42, at 40.

286 N. Peart and D. Holdaway (2000) 'Ethical Guidelines for Health Research with Children' *New Zealand Bioethics Journal* 1 (2): 3-9.

287 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, p 262, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

288 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt and F.M. Giardiello (1996) 'Genetic testing for cancer in children: Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11):1131-8, A.M. Codori, K.L. Zawacki and G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects.' *Am J Med Genet* 116A(2), 117-28.

289 S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26.

290 Rony Duncan 'Holding Your Breath, Predictive Genetic Testing in Young People' Ph.D Thesis, Department of Paediatrics and Public Health, University of Melbourne, July 2005, pp 165-166, accessed on 17 September 2006 from http://eprints.infodiv.unimelb.edu.au/archive/00001621/01/PhD_Thesis_Rony_Emily_Duncan_2005_eprints.pdf).

291 A.M. Codori, G.M. Petersen, P.A. Boyd, J. Brandt and F.M. Giardiello (1996) 'Genetic testing for cancer in children: Short-term psychological effect' *Arch Pediatr Adolesc Med* 150(11):1131-8, A.M. Codori, K.L. Zawacki and G.M. Petersen et al. (2003) 'Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects' *Am J Med Genet* 116A(2), 117-28, and S. Michie, M. Bobrow and T.M. Marteau (2001) 'Predictive genetic testing in children and adults: A study of emotional impact' *J Med Genet* 38(8):519-26.

292 Ellen Wright Clayton (1998) 'Genetic Testing in Children: The Law's Role in Addressing the Challenges Posed by New Technology (Commentary)' *Children's Legal Rights Journal* Vol. 18, No. 2, 39-42, 40.

293 Ellen Wright Clayton (1998) 'Genetic Testing in Children: The Law's Role in Addressing the Challenges Posed by New Technology (Commentary)' *Children's Legal Rights Journal* Vol. 18, No. 2, 39-42, 40. Note that, however, Wright Clayton is concerned with the law's role in addressing genetic testing of children, and she surmises that any argument that the State should not intervene in pre-symptomatic genetic testing because it is not harmful enough to warrant such intervention 'does not resolve the issue because the question is not whether testing children for predisposing mutations should be illegal but whether physicians should accede to parental requests for those tests.'

294 Stephen Robertson and Julian Savulescu (2001) 'Is There a Case in Favour of Genetic Testing of Young Children' *Bioethics* 15:1, 38–42.

295 Stephen Robertson and Julian Savulescu (2001) 'Is There a Case in Favour of Genetic Testing of Young Children' *Bioethics* 15:1, 31.

296 While we previously argued that children are not small adults and research with adults cannot be readily extrapolated to draw conclusions about testing of children, arguments around 'future autonomy' are partly grounded in hypothesising the interests (and rights) of the adult who will develop from the child, and thus we believe that the data is more readily comparable.

297 Susan Hogben and Paula Boddington (2005) 'Policy Recommendations for Carrier Testing and Predictive Testing in Childhood: A Distinction That Makes a Real Difference' *Journal of Genetic Counseling* 14, 4, August, 271–281, 277.

298 Susan Hogben and Paula Boddington (2005) 'Policy Recommendations for Carrier Testing and Predictive Testing in Childhood: A Distinction That Makes a Real Difference' *Journal of Genetic Counseling* 14, 4, August, 271–281, 278.

299 J.K. Mason and G.T. Laurie *Mason and McCall Smith's Law and Medical Ethics* (7th ed.), Oxford University Press, 2006, p 208.