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**Patient and Professional Constructions of  
Familial Hypercholesterolaemia and Heart  
Disease: Testing the Limits of the Geneticisation  
Thesis**

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**Thesis submitted to the University of Nottingham  
for the degree of Doctor of Philosophy**

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## **ABSTRACT**

This thesis provides an empirical investigation of the geneticisation thesis. Geneticisation is one of the most prominent critiques of the social and cultural implications of developments in genetics. It incorporates a set of claims and expectations about the way genetic knowledge and technologies are transforming or will transform ideas about health and illness, and health care practices. This research aims to explore the empirical basis of these claims, by looking at the place of genetic discourses and practices in one specific area. The thesis focuses on familial hypercholesterolaemia (FH), a treatable hereditary cholesterol condition associated with high rates of coronary heart disease (CHD). It asks how much and in what ways patients with FH and professionals involved with the condition construct FH and CHD as genetic conditions.

The thesis draws on three main areas of data - biomedical literature concerning CHD and FH; ethnographic work concerning the activities of HEART UK, the main UK health charity involved with inherited lipid disorders and cholesterol; and interviews with patients with FH and with staff and members of HEART UK. The analysis suggests that FH is not understood or managed within a strong genetic frame, and that neither professionals involved in HEART UK, nor patients with FH, provided or contributed to radically new or geneticised accounts of CHD. In short, the research suggests that geneticisation overstates the transformatory potential of genetics, and that factors such as the availability of effective therapeutics, the sites where care takes place, the disciplines involved, and existing lay and professional models of disease are important for

the construction of a particular field. Furthermore, in arguing that FH is not associated with a strong specific disease identity or community, the analysis questions the notion of biosociality, suggesting that it may be less relevant to some biological states or conditions than to others.

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# CONTENTS

<b>ABSTRACT</b>	2
<b>ACKNOWLEDGEMENTS</b>	4
<b>CONTENTS</b>	6
<b>INDEX OF TABLES AND FIGURES</b>	10
<b>LIST OF ABBREVIATIONS USED IN THE THESIS</b>	11
<b>CHAPTER 1: INTRODUCTION</b>	12
1.1 KEY CONCEPTS	13
1.2 HOW THE IDEAS FOR THIS THESIS CAME ABOUT AND DEVELOPED	18
1.3 WHY FOCUS ON CHD AND FH?	19
1.4 FAMILIAL HYPERCHOLESTEROLAEMIA – THE CLINICAL CONTEXT	20
1.5 RESEARCH QUESTIONS	22
1.6 METHODS	23
1.7 KEY FINDINGS	24
1.8 OVERVIEW OF CHAPTERS	24
<b>CHAPTER 2: LITERATURE REVIEW</b>	28
2.1 GENETICISATION	28
2.2 DISEASE CONSTRUCTIONS	54

<b>CHAPTER 3: RESEARCH DESIGN AND METHODS</b>	87
3.1 INTRODUCTION	87
3.2 CASE SELECTION	89
3.3 ANALYSIS OF BIOMEDICAL LITERATURE	91
3.4 ETHNOGRAPHIC WORK WITH HEART UK	100
3.5 INTERVIEWS WITH HEART UK MEMBERS AND LIPID CLINIC PATIENTS WITH FH	108
3.6 REFLECTIONS ON THE RESEARCH DESIGN AND PROCESS	132
3.7 SUMMARY	138
<b>CHAPTER 4: BIOMEDICAL CONSTRUCTIONS OF CHD AND FH</b>	140
4.1 INTRODUCTION	140
4.2 A BRIEF OVERVIEW OF THE MOLECULAR BIOLOGY OF CHD	141
4.3 MODELS OF CHD IN THE BIOMEDICAL LITERATURE	142
4.4 HEART UK'S CONTRIBUTION TO MODELS OF CHD	158
4.5 HOW TO DEFINE FH?	164
4.6 CHAPTER SUMMARY AND DISCUSSION	177
<b>CHAPTER 5: HOW DOES HEART UK CONSTRUCT THE FIELD?</b>	181
5.1 INTRODUCTION	181



5.2 THE ORGANISATION OF HEART UK AND THE ROLE OF PATIENTS/LAY PEOPLE.	182
5.3 THE FOCUS OF ACTIVITIES	195
5.4 THE AETIOLOGY OF CHD AND HYPERCHOLESTEROLAEMIA	207
5.5 CONSTRUCTIONS OF FH	214
5.6 CHAPTER SUMMARY AND DISCUSSION	223
<b>CHAPTER 6: DEFINING AND EXPLAINING THE PROBLEM</b>	225
6.1 INTRODUCTION	225
6.2 NAMING THE PROBLEM	227
6.3 EXPLAINING FH	228
6.4 EXPLAINING HIGH CHOLESTEROL	230
6.5 LIFESTYLE TALK	237
6.6 TAKING CARE OF ONESELF	242
6.7 EXPLAINING CHD	244
6.8 CONSTRUCTING THE HEREDITARY ASPECT OF FH	248
6.9 CHAPTER SUMMARY AND DISCUSSION	260
<b>CHAPTER 7: LIVING WITH FH</b>	263
7.1 INTRODUCTION	263
7.2 GENETIC RESPONSIBILITY	264
7.3 REPRODUCTIVE DECISION MAKING	265
7.4 PASSING IT ON	270

7.5 LIVING AT RISK OF CHD	273
7.6 TAKING CARE OF CHILDREN WITH FH	276
7.7 TALKING WITH KIN	283
7.8 TALKING WITH OTHER PEOPLE	291
7.9 CHAPTER SUMMARY AND DISCUSSION	293
<b>CHAPTER 8: CONCLUSION</b>	295
8.1 SUMMARY OF FINDINGS	296
8.2 PATIENTS ASSOCIATIONS, KNOWLEDGE AND EXPERTISE	304
8.3 GENETIC RESPONSIBILITY AND BIOSOCIALITY	308
8.4 WHAT CAN BE SAID ABOUT THE GENETICISATION THESIS?	311
8.5 IMPLICATIONS FOR POLICY AND PRACTICE.	319
8.6 QUESTIONS RAISED AND FUTURE RESEARCH	323
8.7 FINAL REFLECTIONS	325
<b>BIBLIOGRAPHY</b>	327
<b>APPENDIX 1: RECENT PAPERS ON GENETICISATION OR CITING LIPPMAN 1991/1992</b>	360
<b>APPENDIX 2: RECRUITMENT DOCUMENTS FOR HEART UK INTERVIEWS</b>	366
<b>APPENDIX 3: INTERVIEW SCHEDULE FOR HEART UK</b>	368

<b>APPENDIX 4: RECRUITMENT DOCUMENTS FOR INTERVIEWS WITH LIPID CLINIC PATIENTS</b>	369
<b>APPENDIX 5: INTERVIEW TOPIC GUIDE FOR LIPID CLINIC PATIENTS</b>	374

## **INDEX OF TABLES**

Table 2.1 Recent empirical studies of geneticisation	47
Table 3.1 Sources of research data for each question	88
Table 3.2 Proposed quota sample	118
Table 3.3 Quota sample, planned and actual sample	122
Table 4.1 Publications of members of the Simon Broome Register group (SB) and Research Committee (R), 2000 onwards.	160
Table 6.1 Route to diagnosis of high cholesterol	250

## **INDEX OF FIGURES**

Figure 4.1 Simon Broome Diagnostic Criteria	167
Figure 5.1 Leaflets for the general public produced by HEART UK	202
Figure 5.2 What Causes CHD?	208

## LIST OF ABBREVIATIONS USED IN THE THESIS

BHA	British Hyperlipidaemia Association
BHF	British Heart Foundation
CHD	Coronary heart disease
CV	Cardiovascular
CVD	Cardiovascular disease
FCH	Familial combined hyperlipidaemia
FDAB	Familial defective apolipoprotein B-100
FH	Familial hypercholesterolaemia
FHA	Family Heart Association
GIG	Genetic Interest Group
HBOC	Hereditary breast/ovarian cancer
HDL	High density lipoprotein
HEART UK	Hyperlipidaemia Education and Research Trust
HGP	Human Genome Project
LDL	Low density lipoprotein
PKD	Polycystic kidney disease
SBHRT	Simon Broome Heart Research Trust
STS	Science and technology studies

## CHAPTER 1: INTRODUCTION

This thesis is about the place of genetic knowledge in understanding and managing common diseases. It is also concerned with changing ideas about expertise. It looks at one condition, familial hypercholesterolemia (FH), an hereditary cholesterol condition associated with high rates of coronary heart disease (CHD). The research focuses on patients with FH and on HEART UK, a health charity formed to support people with hereditary cholesterol problems. The thesis asks how much and in what ways these groups construct FH and CHD as genetic conditions. It also asks in what ways lay people have contributed to particular understandings of FH through their involvement with HEART UK. The aim of the thesis is to consider the claims of the geneticisation thesis (Lippman, 1991, 1992) about the increasing prominence and implications of genetic ways of thinking about health and illness, and to contribute to discussions about the role of lay health groups in the production and dissemination of biomedical knowledge. The study draws on ideas from the fields of medical sociology, science and technology studies (STS) and studies of the public understanding of science.

This introductory chapter will set out the context of this thesis, explain the rationale for the thesis and how this developed, set out the research questions, provide a brief account of the methods and findings, and finish by outlining the structure of the rest of the thesis.

## **1.1 KEY CONCEPTS**

This section provides the context for the thesis by discussing the three main conceptual areas on which it draws: geneticisation, genetic models for common conditions, and expertise and patients' associations.

### **Geneticisation**

In the early 1990s, Abby Lippman (1991, 1992) coined the term 'geneticisation' to describe a process by which both professional and popular discourses about health and illness were increasingly drawing on genetics and genetic differences. Indeed, Lippman argued that genetics had become the dominant discourse about health and illness. Lippman based this notion of geneticisation on observations about both prenatal diagnosis and about the rhetoric surrounding the Human Genome Project at this time, particularly a discourse of 'prediction and prevention' concerning common adult-onset conditions. She argued that the process of geneticisation would influence how health problems are defined and managed; It would affect our health care practices and our values and attitudes, leading to the increasing use of genetic technologies, the stratification of society on genetic lines, changing concepts of normality and abnormality, and an increasing focus on the biological rather than the social conditions associated with health and illness. In short, it would lead, in Lippman's view, to negative social consequences that both reflect and reinforce social inequalities in health. In her view, this process was mainly being driven by geneticists and their allies.

The geneticisation thesis provides a thoroughgoing critique of the socio-political context and consequences of genetic ways of thinking and doing. Lippman's ideas have been widely cited, discussed and enrolled across a range of disciplines, including sociology, anthropology, STS, bioethics, philosophy, and medicine, and in relation to a number of phenomena, such as kinship, ethnicity and race, adult-onset physical diseases, psychiatric illness, and prenatal carrier matching. A large number of analysts have endorsed the idea of geneticisation including some distinguished scholars (see for example Franklin, 2003; Rapp, 1999; Rose, H., 2001). It has also been critiqued on a number of fronts (Condit, 1999; Condit et al., 1998; Hedgecoe, 1998, 1999; Kerr, 2004; Novas & Rose, 2000). One of the main critiques has been that geneticisation allows no place for the agency of lay people to adopt, contribute to, transform or resist genetic discourses (see for example Novas & Rose, 2000; Rose, N., 2001; Rose & Novas, 2004), and this point is reinforced by the few empirical studies of geneticisation that have focussed on patient discourses (Gibbon, 2002; Raz & Atar, 2004).

It is notable that a large proportion of commentators, both critics and proponents of geneticisation, have tended to bracket empirical questions about whether and how genetic discourses and practices are spreading, assuming that this is happening or will happen. They differ, rather, in their analyses of the meaning and implications of this expansion. In neglecting these empirical issues, one can argue that social analysts have not only commented on, but have contributed to a set of expectations about the transformatory nature of genetic knowledge and technologies. There has been little empirical work that

directly engages with the notion of geneticisation, that aims to address whether the changes anticipated about the way health and illness are conceived and managed, are actually materialising. This thesis starts from a position of neither endorsing nor challenging geneticisation, but aims to examine precisely these empirical questions. It contributes to a small and, so far, relatively neglected area of research.

### **Genetic models for common conditions**

STS scholars argue that the development of molecular genetics has led to a shift in conceptions of genetic disease in biomedical discourses (Martin, 1999; Turney & Balmer, 2000). Biomedical models of many common conditions now include the idea that certain genetic variations may increase a person's susceptibility to or predispose them to particular common diseases. Proponents of genetic research envisage a new era of 'preventative medicine', focussed on the prediction and prevention of disease based on genetic information (Turney & Turner, 2000). Visions of a genetic future for managing common conditions are embodied in UK health policy documents such as the genetics White Paper (Cm 5791 - II, 2003: 14):

'Most of the more common diseases, such as heart disease and diabetes also have a genetic component. An individual's susceptibility to these diseases is determined by a combination of genetic factors and environmental factors...Over the next decade, however, it should be possible to identify more genetic factors that increase the likelihood of people developing a given disease. There will then be the option to test people for a predisposition to that disease, or a higher-than-normal risk. Prevention and monitoring services could then be tailored to an individual's needs'.

As this quote suggests, CHD features as a central example in this vision, along with conditions such as diabetes, cancer, schizophrenia and Alzheimer's



disease. Analysts of the social and ethical implications of new genetic knowledge have reinforced this message that common diseases are being reframed in genetic terms (see for example Davison, 1996; Davison et al., 1994; Duster, 2003; Gannett, 1999; Hallowell, 1999, 2000; Helén, 2004; Lemke, 2004; Lippman, 1998; Rose, N., 2001; Sherwin, 2004). Again, it must be noted that there has been little empirical research concerned with the uptake of genetics within biomedical discourses and practices concerning these common conditions. Furthermore, although there is a growing body of research concerning patients' constructions of genetic disease, this has largely been concerned with 'classic' genetic conditions such as Huntington's disease, or with hereditary breast/ovarian cancer. In sum, although there are some notable exceptions (Cox & Starzomski, 2003; Hall, 2004, 2005; Hedgecoe, 2001a, 2002), there has been little research concerned with either professional or patient constructions of genetic susceptibility for common conditions which are managed outside of the genetics clinic, and for which preventative therapies and treatments may be available. This paucity of research seems remarkable in view of the policy relevance of these common conditions and the amount of discussion the genetic models of these conditions have generated among social and ethical analysts.

### **Expertise and patients' associations**

The idea of lay knowledge or expertise, which is based on close experience of a particular phenomenon or disease, has become established within the social science literature, although discussions continue about how to define it and who can be said to have it (Collins & Evans, 2002; Kerr et al., 1998; Lambert

& Rose, 1996; Popay & Williams, 1996; Jasanoff, 2003; Prior, 2003; Rip, 2003; Shaw, 2002; Williams & Popay, 1994; Wynne, 1996a, b, 2003). There has been particular interest in patients' associations and other lay health groups as sites where expertise is challenged and renegotiated. Such groups may aim to influence health policy and research agendas. Recent analyses have highlighted cases where they have influenced the very practice of research and medicine (Epstein, 1995; Heath et al., 2004; Rabeharisoa & Callon, 2002; Rabinow, 1999; Rose & Novas, 2004).

Rabeharisoa & Callon (Rabeharisoa, 2003; Rabeharisoa & Callon, 2002) suggest that patients' associations may be finding formal ways of collectivising experiential knowledge and that a new 'partnership' model of collaboration between patients' associations and professionals has emerged. This model is characterised by the patients' organisation retaining control of their research policy and through the recognition of their unique role in collating and communicating patients' experiential knowledge, which is seen as equal to professional expertise. Other commentators are more sceptical of the potential for lay health groups to contribute to or challenge biomedical science (Petersen, 2002; Stockdale, 1999; Williams, 1989). This suggests that, although contested, patients' associations are a potentially interesting site of lay knowledge and practice, viewed at a collective rather than individual level.

## **1.2 HOW THE IDEAS FOR THIS THESIS CAME ABOUT AND DEVELOPED**

The initial focus for this research was on lay constructions of CHD and FH. As the ideas for the thesis were coming together it seemed from the STS literature that genetic models of common conditions were emerging in biomedical discourses. Furthermore, Adam Hedgecoe (2001a, 2002) had just started to publish the findings of his study, showing the rhetorical strategies whereby biomedical models of schizophrenia and diabetes had become geneticised. Social commentaries on genetic developments seemed to leave little doubt that genetic models of CHD would also emerge (see for example Davison, 1996; Davison et al., 1994; Lippman, 1998; Robert & Smith, 2004; Rose, N., 2001). At the same time, as noted in the previous discussion, the geneticisation thesis said little about the agency of lay people. However, there was growing interest in the contribution of lay people to the production and dissemination of biomedical knowledge, and particularly the role of patients' associations. The initial aims of the thesis were, therefore, to explore the geneticisation thesis by looking at lay constructions of FH and CHD, focussing on individual patients with FH and on a patients' association. This would allow me to examine the degree to which lay people contributed to or resisted the processes of geneticisation of CHD, which I, at that stage, assumed to be occurring in biomedical discourses.

Two key developments changed the shape of the thesis. First, it became apparent that there had been little recent analysis of biomedical constructions

of CHD. It was not at all clear what impact genetics was having in this area. Second, during the course of the first year of this study, the patients' organisation for people with hereditary cholesterol disorders merged with the association for health professionals and scientists working in the field of lipid disorders. The status of this new organisation as a lay health organisation fell into question. My research design shifted to include some analysis of biomedical constructions of CHD and FH, focussing particularly on the professionals involved in the new joint organisation, and to look at the kinds of relationships between biomedical and lay expertise that were embodied within this newly merged organisation.

### **1.3 WHY FOCUS ON CHD AND FH?**

CHD is the single most common cause of death in the UK, accounting for about one in five deaths in men and one in six deaths in women in 2002 (The British Heart Foundation, 2005). It is, therefore, a very important condition in terms of health policy and its financial implications. Furthermore, the condition features as one of the central examples in the vision of preventative medicine based on genetic information outlined above. It is one of the key conditions discussed by the genetics White Paper (Cm 5791 - II, 2003) and is cited as a focus for the nationally-sponsored UK Biobank (The UK Biobank, 2004; The Wellcome Trust, 2003). As noted above, commentators on the potential social and cultural implications of developments in the genetics of common conditions also cite CHD as a key example. In short, CHD is an important common disease for which genetic models seem to be emerging in some quarters.

FH is an hereditary form of raised cholesterol. The condition, itself, is symptomless, but leads to increased risk of early and severe CHD. However, once the raised cholesterol has been identified, it can be successfully treated, substantially reducing the risk of CHD in this group of people (Scientific Steering Committee on Behalf of the Simon Broome Register Group, 1999). The condition is relatively common, thought to affect about one in five hundred people in the UK. FH, therefore, provides an appropriate case through which to examine constructions of genetic susceptibility for a common condition because it provides an example of an established genetic condition that increases susceptibility to CHD, that is relatively common and is treatable.

#### **1.4 FAMILIAL HYPERCHOLESTEROLAEMIA – THE CLINICAL CONTEXT**

FH is characterised as a dominant single gene condition that leads to high blood cholesterol levels. There are two forms of FH, the relatively common heterozygous form and the much rarer and more severe homozygous form<sup>1</sup>. This thesis focuses on heterozygous FH, which will be referred to simply as FH. Studies undertaken before the introduction of effective cholesterol lowering drugs suggest that, without intervention, at least fifty per cent of men and about thirty per cent of women with FH would experience fatal or non-fatal CHD by the age of sixty (Scientific Steering Committee on Behalf of the Simon Broome Register Group, 1999). FH is treated predominantly using a

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<sup>1</sup> People with heterozygous FH have one normal and one defective gene. Homozygous FH results from having two defective genes. Homozygous FH is thought to affect about one in a million people and, untreated, is likely to result in the onset of CHD in the teens or early twenties.

class of cholesterol lowering drugs called statins, combined with dietary and other lifestyle modifications. The condition is currently diagnosed in the UK mainly on the basis of a set of clinical indicators, although DNA-based diagnosis is feasible. Specialist care for people with FH is provided through lipid clinics, which are specialist outpatient clinics. There are currently 135 clinics providing lipid services in the UK (Marks et al., 2003a). Neil et al. (2000) suggest that people who have been diagnosed with FH are rarely cared for exclusively by their general practitioner.

The main health charity involved with FH is HEART UK (the Hyperlipidaemia Education and Research Trust). This organisation was formed in 2002 as a result of the merger of the Family Heart Association (FHA), the main patient-focused charity concerned with inherited high cholesterol, and the British Hyperlipidaemia Association (BHA), the professional association for scientists and health professionals involved with lipid disorders, whose interests included both inherited and acquired conditions. HEART UK must be seen as a kind of hybrid organisation which aims to act as both patients' association and as a professional body. HEART UK is connected to a further group, the Simon Broome Register Group. This is a semi-autonomous research group concerning FH, made up of clinicians and biomedical scientists. It has provided important data on the epidemiology and management of FH. The Simon Broome Register Group is now administered through HEART UK. Furthermore, the group includes several of the leading professionals involved in HEART UK.

## 1.5 RESEARCH QUESTIONS

The aim of this thesis is to consider the claims of the geneticisation thesis by examining constructions of CHD and FH. The overarching question asked by the thesis is:

**To what extent do FH patients and biomedical professionals construct FH and CHD as genetic conditions?**

This question is broken down into three specific areas:

- 1. How are FH and CHD constructed in recent biomedical literature?**
- 2. How does HEART UK construct FH and CHD?**
- 3. How do patients with FH construct FH and CHD?**

Geneticisation must be seen as both *a way of thinking and a way of doing* (Lippman, 1998: 64). The research, therefore, explores how FH and CHD are constructed by looking at different sorts of data. First, it considers *ways of thinking* about CHD and FH, by looking at the way these conditions are explained in explicit accounts of their aetiology provided in the biomedical literature, by HEART UK and by patients with FH. Second, it considers *ways of doing* in relation to CHD and FH, by looking at the way these conditions are constructed through the activities and actions of HEART UK and patients. This means, for example, for HEART UK, looking at who the organisation aims to support, the sorts of groups it collaborates with, the sorts of issues on which it lobbies or advises and the types of research, technologies and health practices it supports or pursues. For patients this means looking at the way

health risks and responsibilities are configured through their talk about their experiences of FH and actions as a result of diagnosis.

The thesis also aims to contribute to discussions about lay expertise and the role of patients' associations in the production of knowledge. The research therefore asks:

**4. What roles do patients play in the construction of discourses about FH and CHD within HEART UK**

This question recognises the unusual hybrid form of HEART UK and is concerned with the degree to which the organisation's activities and discourses can be thought of as being indicative of lay constructions of FH and CHD specifically.

**1.6 METHODS**

This is a qualitative study that draws on three main areas of data (1) analysis of papers published in biomedical journals, including selected recent commentary articles about CHD, the publications of selected professional members of HEART UK and the publications of the Simon Broome Register Group (2) interviews with staff and members of HEART UK, observation of their activities and analysis of the organisation's written materials (3) semi-structured interviews with patients with FH, accessed at a large lipid clinic in the north of England.



## **1.7 KEY FINDINGS**

The thesis challenges the idea of geneticisation in a number of important ways. It suggests that FH is neither understood nor managed within a strong genetic frame. Furthermore, neither health professionals involved with HEART UK nor patients with experience of FH provided or contribute to radically new or geneticised accounts of CHD. In short, the thesis suggests that geneticisation is not evident in this case, where there is a recognised and established hereditary link to CHD. This raises questions about the impact of genetics in other domains of medicine. The thesis also suggests that HEART UK is a highly professionalised organisation. Although patients' experiential knowledge was valued, biomedical matters were largely delegated to biomedical experts. This suggests, in the case considered, a rather less radical role for patients' organisations as an expression of patient expertise than has recently been proposed elsewhere.

## **1.8 OVERVIEW OF CHAPTERS**

This introduction is followed by seven chapters.

Chapter 2, *Literature review*, discusses the notion of geneticisation and reviews the way it has been enrolled since its introduction. It outlines existing sociological work concerning both biomedical and lay constructions of CHD, and the social implications of providing genetic risk information. It then looks at the role of patients' associations in the production and dissemination of biomedical knowledge. The chapter provides the key theoretical concepts drawn on in the thesis and sets out the scope for detailed empirical investigation concerning geneticisation and complex, common diseases.

Chapter 3, *Methodology and methods*, provides an account of the methods adopted and why they were used, including details of how samples were selected and the data analysed. It addresses ethical and practical issues that arose and how the research design and focus developed in response to these issues.

Chapter 4, *Biomedical construction of CHD and FH*, looks at biomedical or ‘expert’ constructions of the aetiology of CHD and FH, focussing on published biomedical literature. It establishes that there are a number of models of CHD and suggests that biomedical professionals involved with HEART UK largely do not focus on genetic models of CHD. It also suggests that there is ambivalence about the clinical utility of genetic information concerning FH. The chapter argues that the different models of CHD can be related to different disciplinary perspectives of the biomedical scientists involved.

Chapter 5, *How does HEART UK construct the field?* focuses on the structure, aims and activities of HEART UK. It argues that the organisation is now professionally dominated and embodies traditional roles concerning expertise. Building on the findings in Chapter 4, it suggests that the organisation does not focus on genetics in relation to CHD risks generally or in relation to FH and that it was characterised by a CHD culture rather than genetic disease culture. The data also reinforce the idea of different disciplinary perspectives and boundary disputes in relation to CHD and FH.

Chapter 6, *Defining and explaining the problem*, looks at the accounts of FH and CHD provided by lipid clinic patients who have FH. Although patients' explanations of FH always included some talk of heredity, the condition was framed in a number of, sometimes, contradictory ways. Their accounts of CHD in general did not draw heavily on genetics and even their explanations of cases of CHD in people with FH were not fixed on the hereditary aspect, but drew on a range of factors. In short, lay models of CHD, embodied by the idea of the 'coronary candidate', appear to be very tenacious, even where specifically hereditary explanations are available.

Chapter 7, *Living with FH*, looks at how the lipid clinic patients with FH frame the condition through their talk about their response to it in their everyday lives. These patients situated FH as part of normal, acceptable, unavoidable, treatable and manageable illness and drew a firm boundary between it and 'serious' genetic diseases. Reproductive decision making was not seen as a relevant theme in relation to FH. Patients' talk revealed a strong sense of responsibility for their offsprings' welfare, but a looser sense of obligation to wider kin. This again suggests that patients did not construct FH through a strongly genetic frame.

Chapter 8, *Discussion and conclusion*, discusses the findings in relation to the key theoretical concerns of the thesis. It concludes that the study provides a fundamental challenge to the geneticisation thesis. The analysis proposes that factors such as the availability of effective therapeutics, the sites where care takes place, the disciplines and technologies involved and existing lay models

of disease have important implications for the construction of a particular disease, suggesting that geneticisation is unlikely to be evident in relation to other common complex conditions. The thesis also concludes that the transformation of expertise reported in relation to other patients' groups is not seen in the area of FH, and suggests that the influence of patients and patients' associations must be related to the state of a particular field and the existing actors within it.

## **CHAPTER 2: LITERATURE REVIEW**

This literature review is divided into two main parts. The first part discusses geneticisation, as this provides the main analytical focus of the thesis. This part introduces the principal arguments of the geneticisation thesis and reviews the way it has been critiqued and enrolled within the academic literature. The second part of the literature review concerns disease constructions, focussing on constructions of CHD and of genetic disease. This provides the context and background for the specific case considered in this thesis.

### **2.1 GENETICISATION**

The geneticisation thesis is one of a number of commentaries that started to emerge in the early 1990s about developments in genetics. This part of the literature review first discusses the main arguments of the geneticisation thesis and their links to the analytical landscape of the time. It then reviews discussions about the notion of geneticisation and how it might best be studied. This is followed by a review of studies that draw on geneticisation, focussing particularly on the recent enrolment of the concept. The aim of this review is to illustrate the wide uptake of geneticisation, discuss the main critiques of the notion and to highlight the main areas that have been discussed or studied under the rubric of geneticisation.

#### **Abby Lippman and the geneticisation thesis**

The geneticisation thesis was introduced by Abby Lippman in two key papers: Lippman (1991) focused on prenatal diagnosis and Lippman (1992) provided a

critique of the Human Genome Project, focusing particularly on a discourse of ‘prediction and prevention’ for common adult-onset conditions. Lippman has reiterated and developed her arguments about geneticisation in several subsequent papers (see for example Lippman, 1993, 1998). Together, these papers provide a wide-ranging critique of the role of genetics in healthcare and make a comprehensive set of claims about genetics and its uses. The following paragraphs are intended to provide a summary of these claims, paraphrasing Lippman’s arguments. These are simply enumerated at this point, without commentary. The claims are discussed in detail in the sections following this.

Geneticisation is defined as:

‘an ongoing process by which differences between individuals are reduced to their DNA codes, with most disorders, behaviors and psychological variations defined, at least in part, as genetic in origin. It refers as well to the process by which interventions employing genetic technologies are adopted to manage problems of health. Through this process, human biology is incorrectly equated with human genetics, implying that the latter acts alone to make us each the organism she or he is’  
(Lippman, 1991: 19).

The central tenets of Lippman’s thesis are:

**1. Genetics is the dominant discourse**

Both professional and popular (mass media) discourses about health and disease are increasingly drawing on genetics. Indeed, this is the *dominant discourse* about health and disease (Lippman, 1991: 15). Common conditions such as cancer, alcoholism and schizophrenia have been reconstructed as genetic diseases.

**2. Genetic discourses are reductionist and deterministic**

They suggest that models of health and disease can be reduced to a set of

(biological) components and that, in the end, genes determine health. This model is inappropriate, even for single gene disorders, where genetic testing neither predicts the severity of the condition, nor shows how it might best be managed and treated.

**3. Society is becoming stratified along genetic lines.**

Geneticisation redefines what are to be understood as significant differences between people, establishing hierarchies among people on the basis of differences in their DNA: ‘Geneticization is the ever growing tendency to distinguish people one from another on the basis of genetics’ (Lippman, 1998: 64).

**4. Geneticisation affects our healthcare practices and our values and attitudes**

Geneticisation is both a discourse and a practice, conditioning how health problems are defined, viewed and managed. It directs the application of intellectual and financial resources and ‘profoundly influences our values and attitudes’ (Lippman, 1991: 18). Specifically it

*a. affects concepts of normality and abnormality*

In the search for susceptibility to common conditions and traits, genetic variations become recast as genetic abnormalities. Similarly, prenatal care practices are ‘restricting concepts of what is “normal”’ (Lippman, 1991: 25).

*b. privatises and individualises health risks and responsibility*

Prenatal screening and diagnosis individualises a woman’s feeling of personal responsibility for her child’s health, just as the idea of genetic susceptibility testing for common adult-onset diseases such as CHD

individualises responsibility to avoid disease through making appropriate behavioural changes<sup>2</sup>.

*c. focuses attention on biological rather than social conditions*

The focus on individual susceptibility and responsibility may detract attention and resources from social programmes to reduce structural risks, thereby increasing social inequalities. Furthermore, opportunities to access and respond to genetic information are likely to reflect and reinforce current social inequalities. Ultimately geneticisation could lead to a ‘coercive model of medicine’, in as much as individuals are positioned as responsible for their health, and so could lead to ‘victim blaming’ of those who do not follow health advice (Lippman, 1992: 1473).

*d. leads to the increased use of genetic technologies in health care.*

Geneticisation means that genetic technologies are ‘applied to diagnose, treat, and categorize conditions previously identified in other ways’ (Lippman, 1998: 64). This means the increasing use of prenatal diagnosis and of predictive or susceptibility testing.

**5. Genetic discourses suggest that genetic research is imperative for future health improvements.**

These discourses suggest that ‘increased understanding of disease and improvement in health will – and can only – be produced by mapping and studying genes’ (Lippman, 1992: 1470).

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<sup>2</sup> It is notable that Lippman’s discussion of genetic prediction and prevention talks only of expected behavioural changes in response to genetic information. The possibility of prophylactic therapies is not discussed.



**6. Geneticisation reflects the power of geneticists to identify and classify health problems.**

A genetic narrative reflects the cultural assumptions of those with the power to name and assign causes. These are mainly the ‘clinical and research geneticists and their colleagues [who] are conditioning how we view, name and propose to manage a whole host of disorders and disabilities’ (Lippman, 1991: 18). The geneticists and their supporters have ‘tremendous power ...for defining how we think of ourselves and others and for determining who will manage us as individuals and as a society (Lippman, 1992: 1474).

In sum, the geneticisation thesis means more than simply an expansion of genetic discourses about disease. It is also more than just a critique, on scientific grounds, of reductionist explanations of disease. Abby Lippman’s geneticisation thesis involves a programmatic critique of the socio-political context and consequences of genetic ways of thinking. It involves a large number of claims concerning the dominance of genetic discourses, changing values and attitudes towards disease and disability, the increasing use of genetic technologies in health care and the powerful role of geneticists in shaping definitions of and responses to disease. These claims have been challenged on both theoretical and empirical grounds and these critiques are reviewed in the following sections. My own research starts from a position of neither endorsing nor challenging geneticisation, but aims to examine the empirical grounds of the claims laid out in the preceding discussion. This position will also be expanded on in the following sections.

## **Background and aims of the geneticisation thesis**

Lippman's analysis is informed by a constructionist model of health and illness. While acknowledging a material reality to disease, Lippman argues that the way health problems are categorised, and causes attributed and studied, is grounded in particular social and cultural assumptions at a particular historical time. Further, her discussion of prenatal testing is informed by feminist arguments about the medicalisation of pregnancy and childbirth. While largely following the conventions of scholarly writing, there is an element of agenda setting and campaigning in these papers. Her aim is not simply to illustrate the social construction of genetic disease categories and a genetic approach to healthcare, but to prioritise a different 'story' about health and illness, concerning the social and structural determinants of health, which are very broadly conceived:

‘Why not seek to change employment, income support, housing and taxation policy that influence the probabilities for illness in a population instead of – or at least in addition to- lobbying for ‘lifestyle’ modifications?’  
(Lippman, 1992: 1473).

This argument exemplifies the more general and radical critique of public health practices, labelled the ‘ecological perspective’ (Lupton, 1995), demonstrating that the geneticisation thesis is part of a much wider debate about the construction and management of health and illness.

Lippman's focus reflects her biography, combining academic interests in the areas of epidemiology, bioethics and social studies of medicine, with a long-time commitment to health activism, particularly relating to women's health. It is also important to point out the context of Lippman's original papers,

published in 1991 and 1992. They were partly intended to counteract the hyperbole surrounding the Human Genome Project (HGP) and to provide a broadside attack on ‘mainstream’ bioethical analysis of the time, with its focus on individual choice and autonomy. Lippman’s papers can be seen as a call for more fundamental analyses of the social and ethical implications of developments in genetics. Regardless of one’s views about the specific claims of the geneticisation thesis, these aims were both timely and valuable. The geneticisation thesis is one of a number of similar analyses that emerged in the first half of the 1990s of contemporary developments in genetics, including Duster (1990), Hubbard and Wald (1993), and Nelkin and Lindee (1995). While these books have different foci and employ different terms, they share many of the key elements of the geneticisation thesis. They demonstrate that Lippman’s papers contributed to a wider critique or unease about the (purported) growing prominence of genetic discourses and practices around this time.

More widely than this, geneticisation resonates with a number of themes that are seen as characteristic of late or reflexive modernity (Beck, 1992; Giddens, 1991) or of neoliberal government (Rose, 1996), in which risk, choice and individual agency and responsibility are central organising ideas. Beck (Beck, 1992; Beck & Beck-Gernsheim, 2001) argues that contemporary society is characterised by ‘individualization’, where traditional social categories no longer determine biography; individuals become the agents of their own life-plans and identity (Beck, 1992; Beck & Beck-Gernsheim, 2001; Giddens, 1991). Contemporary society is also a ‘risk culture’ (Giddens, 1991: 3),

preoccupied with the future consequences of actions in the present. This finds expression through the prominence of risk assessment and risk management across a range of political programmes (Murphy, 2000). Drawing these two themes together, Rose (1996: 58) argues that in advanced liberal society, collective forms of risk management, such as social insurance, have given way to the 'privatization of risk management', in which 'the citizen is enjoined to bring the future into the present, and is educated in the ways of calculating the future consequences of actions'. Indeed, a calculative and prudent relation to the future becomes an individual obligation.

The field of health is exemplary of these rationales of risk and individual agency. Contemporary health prevention discourses are premised on the notion that risk factors can be identified and illness avoided, mainly through the actions of individuals. In the construction of risk factors, through the techniques of epidemiology, there is a tendency to see certain risk factors as more manageable than others, privileging those that focus at an individual level. Although the evidence for these factors may be contingent and uncertain, this uncertainty is lost as the evidence is translated into solid guides for action concerning 'lifestyle' through the discourses of public health. (Petersen & Lupton, 1996). Under the dominant discourse of 'healthism' (Crawford, 1980), health becomes the result of rational choice and individual responsibility (Greco, 1993; Lupton, 1995). All of this suggests that the arguments put forward by Lippman about genetics are not specific to genetics. Claims that geneticisation is reductionist, privatises and individualises health risks and responsibilities, and may lead to victim blaming, must be set within

the context of wider rationales relating to the organisation of health care and society. These arguments will be expanded on particularly in relation to CHD in the second part of this chapter.

### **Uptake of geneticisation**

In the decade and a half since Lippman's original publications, geneticisation has been widely cited, enrolled and discussed across a range of disciplines, including sociology, anthropology, communication studies, STS, bioethics, philosophy and medicine, and in connection with a range of phenomena and conditions, including ethnicity/race, kinship, a number of adult-onset physical diseases, psychiatric illness, sexuality, smoking tobacco, genetic enhancements, pharmacogenetics, prenatal screening and prenatal carrier matching. Citation data give some indication of the wide dissemination, although not necessarily support, of Lippman's ideas. The ISI Web of Science (WOS) database lists some 130 references that cite either Lippman 1991 or 1992. Searching WOS and the CSA Applied Social Sciences Index and Abstracts (ASSIA) databases for 'geneticization' or 'geneticisation' resulted in a total of 41 references<sup>3</sup>. These searches are obviously not exhaustive. In particular, books and book chapters are likely to be missing. In addition, the term geneticiz/sation is sometimes used as a generic term, without reference to Lippman. Such references are difficult to capture systematically. It should be noted that the review includes only those papers that focus on geneticisation or refer to Lippman. There are other studies of the social construction of genetic diseases, some of which are discussed in the second part of this chapter. The

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<sup>3</sup> Searches undertaken on 19.4.05.

following sections first discuss the main critiques and commentaries on the notion of geneticisation, then the main proponents of geneticisation up to the early 2000s. This is followed by a systematic review of the recent enrolment of geneticisation, in order to explore how far this concept has travelled.

*Commentary on the notion of geneticisation*

A number of aspects of the thesis have been discussed. Some analysts have critiqued the empirical basis of the thesis, contesting the increasing dominance of genetic discourses (Condit, 1999; Condit et al., 1998). Others do not necessarily contest the idea of increasing focus on genetic discourses, but have discussed what sort of concept geneticisation is, how it is defined and how it might be studied (Cox & Starzomski, 2003; Hedgecoe, 1998, 1999, 2001b, 2002, 2004a; Hoedemaekers, 2001; Hoedemaekers & ten Have, 1998; Kerr, 2004; ten Have, 2000), or have contested the specific claims of the thesis, seeing the social, cultural or political implications of genetic knowledge in a different way (Novas & Rose, 2000; Rose, N., 2001; Rose & Novas, 2004).

Condit's analyses are unusual in so far as they actually challenge claims about the increasing significance of genetic discourses, based on her studies of popular media discourses and the audience reception of metaphors. In fact, these are mainly a critique of Nelkin and Lindee's (1995) analysis of popular writing and images associated with genetics (see Hedgecoe, 1998 for a more detailed review).

Hedgecoe (1998, 1999, 2001a, b, 2002, 2003a, 2004a) is probably the most prolific commentator on the notion of geneticisation as well as an important contributor to empirical work in this area. His critique of geneticisation (Hedgecoe, 1998, 1999, 2001b) can be situated in his own background in bioethics and can be seen as a call for a move to empirical research rather than bioethical analysis concerning geneticisation. In principle, he is critical of the use of geneticisation in bioethical analyses, focussing his attention on two particular studies by Hoedemaekers & ten Have (1998) and Sherwin & Simpson (1999). He argues that geneticisation as formulated by Lippman and enrolled in these studies is an inherently critical concept, in other words that the implementation of genetic technologies can only be seen as negative; its use as an analytical tool for assessing the ethics of genetic technologies leads to ‘moral circularity’:

‘since geneticization is inherently negative towards genetic technologies, using it to assess the ethics of genetic technologies results in the rather predictable conclusion that such technologies have unwelcome, unethical effects’  
(Hedgecoe, 2002: 8).

Hedgecoe (1998) argues that the failings of geneticisation as an analytical tool stem from its origins in medicalisation, and that it is open to the same criticisms levelled against the original formulation of medicalisation. In parallel with these earlier discussions, he argues that the inherently negative framing of geneticisation prevents ‘a balanced discussion of the pros and cons and an accurate picture of how the process of geneticisation takes place’ and proposes that what is required is: ‘a more neutral attitude’ (Hedgecoe, 1999:17) and ‘convincing empirical evidence rather than theory-derived polemic’ (Hedgecoe, 1998: 235). On this basis, he makes two related suggestions for the

analysis of geneticisation: (1) he suggests a ‘stripped down’ definition, which is ‘geneticisation takes place when a condition is linked to a specific stretch of DNA’ (Hedgecoe, 2002: 8) and (2) he calls for ‘detailed empirical research on how geneticisation is taking place, carried out in as many different contexts as possible’ (ibid: 23).

Hedgecoe (2001a, 2002, 2003a) employs his stripped down definition in his own studies of scientific texts concerning schizophrenia, diabetes and cystic fibrosis. These studies analyse a small number of review papers written by influential scientists to show how the rhetorical features of their writing work to construct particular arguments and models, in this case a focus on genetics. Hedgecoe’s analyses are very interesting from an STS perspective, concerning the construction of scientific models and arguments within published papers. Yet the stripped-down definition and the method he uses suggests that is it enough that some scientists frame a condition in a genetic way in order for it to be considered geneticised. Hedgecoe’s definition includes no explicit references to whether such genetic models are dominant or widely accepted, or the possible repercussions of such models.

My discussion of Hedgecoe’s work illustrates the difficulty of deciding whether and when an area could be considered to be geneticised. As Cox & Starzomski (2003) comment, how we research geneticisation is related to how we understand the concept. While Hedgecoe’s work has been important in setting the agenda for empirical investigation of geneticisation and for illustrating one way of studying this area, I am not convinced of the need for



his stripped-down, ethically neutral definition of geneticisation. One might agree that geneticisation is a bad thing if the empirical reality matched up to the expectations spelt out in Lippman's thesis. However, as indicated earlier in this chapter, my interest at this stage is less in the normative questions, and more concerned with the empirical questions about *whether* and *how* genetic knowledge and technologies are changing disease concepts, health-related practices, social relations and values. Lippman's construction of geneticisation is useful for keeping these areas in view, and her wider claims can be supported or challenged through empirical studies. In my view, Lippman's work can be used to set an agenda for empirical work. I intend to organise my own analysis around what I have interpreted to be the key tenets of Lippman's thesis. To recap, these are that: genetics is the dominant discourse; genetic discourses are reductionist and deterministic; society is becoming stratified along genetic lines; genetic discourses are affecting our values, attitudes and health care practices; genetic research is seen as imperative; and geneticists have great influence in how health problems are classified and managed. My aim is to examine these statements through empirical investigation.

Such empirical enquiry might take place at a number of levels<sup>4</sup>:

1. popular culture e.g. books, films, images, media
2. scientific discourse and practice e.g. written texts, conferences, scientific practices, development and uptake of particular technologies, institutional changes

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<sup>4</sup> ten Have and Hoedemaekers (Hoedemaekers, 2001; Hoedemaekers & ten Have, 1998; ten Have, 2000) also suggest a number of levels at which geneticisation could be studied including conceptual, institutional, doctor-patient, cultural and philosophical. These should be seen as conceptual or analytical levels rather than practical or empirical levels for investigation.

3. social/health/science policy discourse and practice e.g. analysis of policy initiatives, documents and processes, funding patterns
4. clinical discourse and practice e.g. technologies and practices adopted, doctor-patient interaction
5. lay discourses and practices e.g. patients, prospective parents

Research at these different levels could provide evidence about changing definitions, practices, values and attitudes concerning health and disease and about the relative influence of geneticists.

It is notable that Hedgecoe talks of the need to elucidate *how* not *whether* a process of geneticisation is taking place, demonstrating his commitment to the concept. Kerr (2004) has questioned this analytical stance in her thoroughgoing critique of Hedgecoe's (2003a) work on cystic fibrosis. Kerr claims that Hedgecoe's analytical focus on geneticisation means that genetic reductionism is foregrounded at the expense of ambiguity and uncertainty, criticises his methodological focus on a small number of papers, and argues that, overall, this provides an ahistorical and inflexible model. She concludes that geneticisation is of limited value when applied to the social construction of genetic disease. Since it can be argued that many diseases are being reframed as genetic, it is not clear what Kerr means by 'genetic disease', or whether she thinks geneticisation may have utility elsewhere. Although Kerr concludes by enjoining us to 'give up' on geneticisation, her paper seems to be more a critique of Hedgecoe's particular methods and analysis.

Hoedemaekers (2001) and ten Have (2000) have also taken issue with Hedgecoe. They regard geneticisation, like medicalisation, as an heuristic tool that helps bring different moral perspectives into view. Reflecting Lippman's own arguments, they suggest that it draws attention away from the dominant ethical debate at the level of individual decision-making to the wider socio-ethical issues. Pointedly, and mainly in response to Hedgecoe's critique of their work (see Hedgecoe, 1999; Hoedemaekers & ten Have, 1998), they claim that geneticisation should not be seen as a sociological explanation of the reality of scientific and everyday life, but rather as a 'philosophical interpretation of the self-understanding of today's human life and culture' (ten Have, 2000: 298), which is not necessarily demonstrable through empirical research. Their exchange with Hedgecoe is part of a larger debate about the relationship between bioethics and social science (Hedgecoe, 2001b), which goes beyond the scope of this review. Nevertheless, it would seem that both ethicists and social scientists can fruitfully engage with geneticisation, regardless of any boundary work between these disciplines (Gieryn, 1983; Hedgecoe, 2001b)

Novas and Rose (Novas & Rose, 2000; Rose, N., 2001; Rose & Novas, 2004) make several important points, suggesting, overall, a more complex and nuanced picture of the role and implications of new genetic knowledge. First, they argue that geneticisation ultimately implies the subjection and control of individuals and groups. They disagree with this construction of patients at genetic risk as passive, arguing that:

'they are increasingly demanding control over the practices linked to their own health, seeking multiple forms of expert and non-expert

advice in devising their life strategies, and asking of medics that they act as the servants and not the masters of this process' (Novas & Rose, 2000: 489).

Second, they argue that genetic information is both individualising and collectivising. It is individualising because of an expectation of 'genetic prudence', the responsibility to manage oneself in the light of knowledge about one's future. However, it is also collectivising in two ways, through locating individuals in a network of relations and through increasing 'biosociality' (Rabinow, 1992):

'Choices about marriage, procreation, financial planning, inheritance, career and much more are made in a web of entanglements involving actual and potential kin, employers, partners and children. And 'at risk' individuals are joining into groups and organizations, not merely demanding public provision and rights, but making their own claims on the deployment of biomedical technologies and the direction of biomedical research' (Rose, N., 2001: 19).

Third, they argue that genetic identity must fit within a multitude of other identity claims and that it is rarely hegemonic; it will not necessarily be the dominant aspect of identity in administrative or regulatory systems such as insurance or legal cases. In essence, this different vision is based on very different conceptions of power relations. Rose draws on the notion of 'pastoral power', a form of relational power, which is diffuse and multi-directional. It cannot be thought of as organised or administered by 'the state', however widely this is conceived.

Several observations can be made about Novas and Rose's analysis. Franklin (2003: 74) also comments on the agency of lay people/patients in the face of genetic information, arguing that ethnographic accounts of the new genetics suggest:

‘There is a lot of picking and choosing going on at the level of which information is accepted as useful knowledge, what kinds of authority are relied upon, and how individual decisions are reached’.

As is shown later, the agency of patients is one of the recurrent themes in discussions about geneticisation, echoing criticisms of the medicalisation thesis (Lupton, 1997). The idea that genetic knowledge is collectivising is interesting, but seems to be based on a particular model of genetic disease as a classic single gene condition. Indeed, Novas and Rose often use the example of Huntington’s Disease. It is difficult to see whether this web of connections applies in the same way to other cases. For example, some visions of genetic susceptibility testing, based on multiple or minor genetic variations, may imply genetic prudence for the conduct of one’s own life, but are unlikely to have implications for choice of partner or reproductive decisions. Finally, it is notable that ideas about increasing biosociality and changing relations concerning expertise are integral to Novas and Rose’s critique of geneticisation. These notions will be discussed in more detail in the second part of this chapter.

#### *Proponents of geneticisation*

Key proponents of geneticisation provide an indication of the range of its uptake. They include Hoedemaker and ten Have (1998), Hallowell (1999, 2000), Sherwin and Simpson (1999) and Hedgecoe (2001a, 2002, 2003a), all of whom cite Lippman and use geneticisation as a frame for their studies or discussions concerning prenatal screening programmes for  $\beta$  thalassaemia, BRCA testing for breast cancer and biomedical constructions of diabetes, schizophrenia and cystic fibrosis. Rapp (1999) endorses the idea of

geneticisation as an ideology or worldview in her detailed ethnography of prenatal screening. Gannett (1999) takes geneticisation as a starting point for her philosophical analysis of the meaning of 'cause'.

Authors who employ geneticisation as a generic term, without reference to Lippman, include Franklin (n.d., 2003), H. Rose (2001) and Petersen (1998). Franklin (n.d.) refers to the 'geneticisation of medicine, reproduction, kinship and identity'. Franklin (2003: 72) talks of the 'geneticization of reproductive choice' and discusses the implications of geneticisation for kinship, drawing on the ethnographic work of Finkler (2000) and Rapp (1999). H. Rose's (2001: 8) work on the Icelandic database, talks of the 'geneticisation of clinical disease entities' and the 'cultural geneticisation of disease' (ibid: 10), which seems to refer to the turn to genetic explanations in aetiological models and the widening uptake of genetic models in the lay population. Petersen (1998: 68) refers to the 'geneticization of society' and 'the 'geneticization' of health and illness' (ibid: 69), and his paper implies increasing scientific and popular interest in the genetic basis of a range of diseases, behaviours and attributes and changing notions of normality and health as a result of developments in genetics.

In sum, these discussions illustrate that a number of authors, including some highly distinguished scholars, have adopted the notion of geneticisation across a range of topics. It is not always obvious what these authors mean when they enrol the concept of geneticisation and how much of Lippman's original conception they are evoking.

### *Recent enrolment of geneticisation*

This section provides a comprehensive review of recent papers that either cite Lippman (1991) or Lippman (1992) or were identified through a key word search on geneticisation/geneticization. The details of the searches undertaken were outlined earlier in the chapter (see page 36). This analysis was undertaken in order to explore systematically the ways in which geneticisation has recently been enrolled, what kinds of empirical work has been undertaken and the degree to which the concept is being endorsed or critiqued. The searches found 14 papers that focused on geneticisation in the last three years, 2002-2004. These constitute more than one third of the total 41 papers identified. Furthermore, this sample contains the majority of the empirical papers found. Details of these sample papers are provided in appendix 1. To typify their stance on geneticisation, six of these papers explore and critique the concept through their analyses (Gibbon, 2002; Hall, 2004; Kerr, 2004; Raz & Atar, 2004; Shaw, 2003; Wilcox, 2003), five papers explore and support the notion through their analyses (Cox & Starzomski, 2003; Hedgecoe, 2002, 2003a, 2004a; Melendro-Oliver, 2004) and three of the papers enrol the idea of geneticisation without questioning it (de facto geneticisation) (Chadwick & Aindow, 2004; Ellison & Jones, 2002; ten Have, 2003).

Nine of the papers report on empirical studies, providing data on a number of levels. Key features of these papers are summarised in Table 2.1 below.

**Table 2.1 Recent empirical studies of geneticisation**

<b>Paper</b>	<b>Area/illness</b>	<b>Level of data</b>
Wilcox (2003)	sexuality	Media discourses
Melendro-Oliver (2004)	Complex diseases & traits	Media & scientific discourses
Hedgecoe (2002)	Diabetes	Scientific discourse
Hedgecoe (2003a)	Cystic fibrosis	Scientific discourse
Shaw (2003)	Dysmorphology	Clinical practice
Gibbon (2002)	Breast cancer	Clinical & patient discourses
Cox & Starzomski (2003)	Polycystic kidney disease	Clinical & patient discourses
Hall (2004)	Heart disease	Clinical & patient discourses
Raz & Atar (2004)	Spouse selection & prenatal screening	Lay/patient discourses

Wilcox (2003) questions whether **media coverage** of biological research on sexuality should necessarily be interpreted as geneticised. Hedgecoe and Melendro-Oliver are concerned with **scientific constructions** of genetic disease. As already discussed, Hedgecoe's (2001a, 2002, 2003a) work, including a slightly earlier paper, shows how common conditions such as diabetes and schizophrenia can be constructed within biomedical literature in such a way as to prioritise genetic elements at the same time as downplaying other elements in aetiological models. Melandro-Oliver (2004) provides a broad-brush analysis of the concept of genetic disease, drawing on biomedical and media texts. She argues that common diseases such as diabetes, asthma and hypertension are being redefined in genetic terms, and indeed that genetic disease is being reframed from a disease category to a new explanatory model for all types of diseases. She concludes that the last decades have not seen an



end of reductionism and genetic determinism in scientific discourses, despite the findings of the HGP, which might contradict these (for example the much lower than expected number of genes). Although this paper makes a number of interesting points, the lack of discussion of the method and the selection of data severely detracts from the author's claims about the dominance of this new genetic disease model.

Cox & Starzomski (2003), Hall (2004), Hedgecoe (2002, 2003a) and Shaw (2003) all raise questions about the utility of genetic information and technologies in **clinical practice**. This is an area that Kerr (2000) has also commented on, in an earlier paper. Hall (2004) provides the only research relating specifically to CHD, undertaken with health professionals and patients at a coronary unit. He reports that the health professionals say they are likely to foreground lifestyle issues in consultation with their patients, because of a concern about fatalism, whereas patients would have liked genetics to be acknowledged. Davison et al. (1989, 1992) made similar observations, at a time almost predating the HGP, about the lifestyle-centred focus of clinicians and health educators compared with a lay interest in heredity and heart trouble. The health professionals' lack of focus on genetics reported by Hall may reflect the current paucity of genetic technologies and treatments in this area, but Hall provides no discussion on this. Nevertheless, this work suggests that clinical discourses in the area of coronary care have not changed significantly in the last fifteen years. It is not clear the extent to which the patients' discourses reflect a geneticised model, or a more long-standing hereditarian discourse about CHD reframed in the language of genetics. Cox & Starzomski's (2003:

161) study of autosomal dominant polycystic kidney disease (PKD) found that ‘recent advances in genetic knowledge and techniques [have] had a minimal impact on the clinical management and social construction of PKD’. The authors propose a number of factors that might mitigate geneticisation such as the management of PKD along with all other kidney disease, the lack of a PKD culture, and the availability of non-genetic diagnostic techniques. This discussion of mitigating factors seems to indicate that Cox & Starzomski (2003) are committed to the idea of geneticisation, although their findings could be interpreted as detracting from the notion.

In addition to Hall’s work, discussed above, there were two other papers that focus on **patient discourses**. Gibbon (2002) and Raz & Atar (2004) discuss the agency of patients in the two very different settings of breast cancer genetics and community genetic services for prenatal carrier screening and matching. Both argue that patients were not passive recipients of service, and that analyses must take account of patients’ expectations and how they respond to and use such services.

In sum these papers suggest that genetic models for common conditions are emerging at the level of scientific discourse, although it is not clear that they represent the dominant constructions. However, there are questions about the current utility of genetic information and techniques in clinical practice and the uptake of genetic discourses by clinicians, as well as questions about the passive position attributed to patients within the geneticisation thesis. There

have apparently not been any empirical analyses in this time period that focus on policy discourses and practices.

The papers discussed so far have geneticisation as their analytical focus. To indicate the wider uptake of Lippman's ideas, there were a further 15 papers in the last year, 2004, that cited either Lippman 1991 or 1992 (see appendix 1 for details). It is instructive to see which of Lippman's ideas are being enrolled. The argument with which she was most frequently associated was that there was increasing uptake of reductionist and deterministic genetic models of health and illness. Mostly this referred to popular culture, lay discourse or society in general, but occasionally scientific discourse or medical practice were specified. There were a much smaller number of references to arguments about the individualisation of responsibility, ideas about normality, new genetic identities or the diversion of resources. Crudely, these papers can be divided into nine that enrol Lippman's ideas without questioning them (Baylis & Robert, 2004; Kirby, 2004; Robert & Smith, 2004; Santos & Maio, 2004; Schubert, 2004; Sherwin, 2004; Shields et al., 2004; Surbone, 2004; van Delden et al., 2004) and six papers that critique some aspects of her analysis (Ceccarelli, 2004; Condit, 2004; Greco, 2004; Helén, 2004; Koch, 2004; Lemke, 2004). The critiques they make have largely been covered in the preceding discussions.

Overall, this review suggests that geneticisation is often used to imply simply an increase in genetic discourses. Furthermore there is frequently limited specificity as to the location of these discourses. It also suggests that, more

often than not, Lippman's arguments are enrolled without question, implying a de facto geneticisation. In as much as references to Lippman continue to increase and her work is often enrolled uncritically, we do not appear to be 'giving up on geneticisation' (Kerr, 2004).

*The construction of genetic developments in social science and bioethical analyses*

There is one final observation to make about the papers reviewed in this part of the chapter. Regardless of whether they endorse or critique Lippman's work and geneticisation, a large proportion of the papers construct genetic developments as leading to the wide-scale transformation of knowledge and practice concerning health and illness. Proponents of geneticisation including Duster (2003), Gannett (1999), Hallowell (1999, 2000) and Sherwin (2004) as well as critics including Helén, (2004), Lemke (2004) and Rose, N. (2001) reinforce the message that common diseases are being reframed in genetic terms. Heart disease is almost always cited as a key exemplar, along with cancer, schizophrenia, Alzheimer's, and diabetes. For example, Rose, a key critic of geneticisation, asserts that it is now 'routine for doctors as well as geneticists to consider that any individual's vulnerability to any disease has a genetic component' (Rose, N., 2001: 11-12) and that tests for genetic variations associated with Alzheimer's, breast cancer and certain types of heart disease are increasingly in development. Robert & Smith (2004), recent proponents of the idea of geneticisation, assert that governments and funding agencies are focussed on 'genetic predisposition to cancer and heart disease' rather than the broad determinants of health. There is little doubt, then, for these

commentators that conditions such as heart disease are being reconstructed as genetic at a number of levels.

Given the fairly widespread assertion that complex common conditions are being reconstructed as genetic, there appears to have been astonishingly little systematic empirical research into the recent scientific, policy and social constructions of such conditions, particularly those managed outside of clinical genetics. Although a number of papers included in this review are concerned with prenatal screening or with hereditary breast/ovarian cancer, Hedgecoe (2001a, 2002), Cox & Starzomski (2003) and Hall (2004) provide the only research that focuses on the construction of common conditions outside of the genetics clinic.

### **Geneticisation: Summary**

This review has argued that the geneticisation thesis should be understood as a programmatic critique of the socio-political context and consequences of genetic discourses about health and illness. It encompasses claims about the widening uptake of (reductionist) genetic models, changing definitions of normality and abnormality, shifting attributions of health risks and responsibilities, the increasing use of genetic technologies in health care, and the role of particular actors in bringing about these changes. While geneticisation contributed to a growing unease in the early 1990s about developments in genetics and their dissemination, its foundations are found in a radical critique of public health practices, whose concerns extend beyond the domain of genetics. Further than this, its themes resonate with the wider

characteristics of contemporary life, whether characterised as late modernity or neoliberal society.

Critiques of geneticisation have challenged the thesis in a number of ways, questioning the uptake and dominance of genetics, challenging the model of power relations it embodies, and questioning its use as an analytical tool. Nevertheless, Abby Lippman's arguments have been widely enrolled across a large number of disciplines and in connection with a wide range of disease conditions and phenomena. It is not always clear what aspect of geneticisation is being enrolled, but it often seems to mean simply an expansion of genetic discourses or criticism of deterministic and reductionistic models, rather than the programmatic critique proposed by Lippman. In view of the wide enrolment and discussion of Lippman's ideas, there have been surprisingly few empirical studies of geneticisation, and even fewer that focus on common conditions outside of the genetics clinic. With some notable exceptions, the papers reviewed in this chapter often bracketed empirical questions about *whether* and *how* genetic discourses and practices are spreading, tending to assume a de facto expansion of genetic ways of thinking and doing.

This thesis is concerned with precisely these empirical questions about whether and how genetic knowledge and technologies are changing disease concepts, health practices, social relations and values. The review has identified a paucity of empirical work concerning common conditions. This study of FH, a genetic condition associated with susceptibility to CHD, contributes to this hitherto relatively neglected area. I have suggested that empirical study of

geneticisation can take place at a number of different levels. This study bridges several of these levels, looking at written scientific, health practitioners' and lay peoples' discourses about CHD and FH. In contrast to much of the literature reviewed, I have explicitly defined geneticisation and detailed the set of claims that I think geneticisation incorporates. These will be used to orient my analysis.

## **2.2 DISEASE CONSTRUCTIONS**

This part of the literature review is divided into three main sections concerning biomedical constructions of CHD and genetic disease, lay constructions of CHD and genetic disease, and the contribution of patients' organisations to disease constructions. It charts the rise of a 'risk factor' model for CHD within biomedical discourses and discusses the more recent emergence of a 'genetic' model for common conditions including CHD. It then discusses lay constructions of CHD, showing the place of genetics in lay models of CHD, and the possible social implications of framing a condition as genetic. This section includes discussion of the (limited) existing research concerning lay constructions of FH. The final section reviews recent discussions that have problematised the distinction between lay and professional knowledge. These arguments are then related to discussions about the rise of lay health groups and their role in knowledge production.

Like Lippman's notion of geneticisation, the discussion here, and in this thesis overall, is premised on a constructionist view of disease aetiology and categories. There are a number of different versions of social constructionism (Bartley, 1990; Brown, 1995a), differing in the role attributed to particular

actors, structural factors and ‘non-human’ entities such as biology or technology, but they would all recognise that what are understood to be the medical ‘facts’ at any particular time are shaped by social and cultural factors. As Yoxen (1982: 144) argued in relation to the construction of genetic disease, the idea that biomedical knowledge is socially determined:

‘does not demand, as some might suppose, that we regard medical conditions as mere artefacts of their social context without organic cause... One can claim... that many of the phenomena of genetic disease are grounded in material reality, whilst at the same time asking why we isolate or delineate certain phenomena for analysis, why we say that they constitute diseases and why we seek to explain their nature and cause in genetic terms’.

My own position is similar to Lippman’s and Yoxen’s, in as much as it recognises that there is a material reality to disease, but asks why certain conditions come to be recognised at a particular time, or become categorised and understood in particular ways. The thesis is concerned with the circumstances under which a condition might be understood as genetic.

### **Biomedical constructions**

#### *Biomedical constructions of CHD*

Aronowitz’s (1998) account of the history of CHD suggests that during the twentieth century angina pectoris became redefined from a condition of chest pain, which was not linked to a specific and localised pathology, to a condition associated with coronary arteries which was linked to specific anatomical and physiological changes. Aronowitz argues that in the first half of the century angina was understood to be related to individual predisposition, and a range of social influences, in particular the pace of modern life. A holistic approach to treatment was advocated which might include drugs, but also aimed to help



patients adjust to or counteract the characteristics of modern life or a particular constitution. Linking CHD to the conditions of modern life was congruent with claims in the mid century that the incidence of the disease had increased dramatically since the First World War. However, observers then and more recently have questioned whether this was a new epidemic of CHD or reflected the emergence of new diagnostic categories such as coronary thrombosis (Aronowitz, 1998; Bartley, 1985). By the early fifties, angina had become largely associated with coronary thrombosis, but was still seen as a chronic degenerative disease connected to ageing, that was not amenable to specific preventative measures.

The second half of the century saw the emergence of the ‘risk factor’ approach to CHD. Indeed, according to Aronowitz (1998), the very term ‘risk factor’ was first used in relation to CHD<sup>5</sup>. By the early 1960s the accepted model of CHD linked specific behaviours such as smoking, measurable physiological characteristics such as high blood pressure, or a family history of CHD, with increased risk of CHD. Aronowitz relates the ascendance of this model to a number of factors including the increasing focus of epidemiologists on chronic disease (the epidemiological transition) and a shift to seeing chronic disease as a result of specific mechanisms that may be preventable, the development of new laboratory tests and of new statistical methods. As noted earlier in this chapter, risk has come to be a central cultural construct in western society (Beck, 1992; Giddens, 1991; Lupton, 1993). The rise of the risk factor approach in CHD can be seen as part of a general increase in focus on risk

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<sup>5</sup> Aronowitz (1998: 119) suggests that the first use of the term ‘risk factor’ was in 1961 in a report on the Framingham Heart Study.

within medical discourse, which has been attributed to a similar set of factors (Skolbekken, 1995).

The new model of CHD is multi-causal, yet as both Aronowitz (1998) and Petersen and Lupton (1996) argue, the risk factor approach tends to privilege individual level risk factors. This can be in part attributed to nature of risk factor modelling:

‘Putative risk factors need to meet certain conditions. They need to be measurable and specific characteristics of the individuals in order to fit into the risk equations that express the results of epidemiological trials. While pack-years of smoking could be entered easily into risk factor formulas, the role of farm subsidies to tobacco growers or marketing of high-fat foods are not so readily modeled’.  
(Aronowitz, 1998: 133)

Earlier ideas relating CHD to the features of modern life were reformulated in risk factor terms. For example, the Type A hypothesis emerged in the 1950s, which linked individual proneness to CHD to a set of behaviours and characteristics particularly associated with white middle-class men, concerning ambition, competitiveness and a preoccupation with deadlines. Riska (2000, 2002) argues that ‘Type A behaviour’ and the ideas of ‘Type A personality’ and of ‘hardiness’ by which it was successively replaced provided the antecedents to the construction of ‘stress’ as a risk factor for a number of chronic diseases. She argues that these psychosocial explanations of CHD provide individualistic frameworks for explaining disease, although they are derived from social categories, particularly class and gender.

There has been continued discussion by epidemiologists and others concerning the limitations of risk factor models in explaining the distribution of disease

and their inability to include factors at levels above the individual. Indeed, as the first part of this review argued, Lippman's geneticisation thesis can be seen as belonging to this critique. In the UK, alternatives to these individual-focussed models can be found in the programmes of epidemiological research concerned with health inequalities (see for example Davey Smith, 2003; Marmot & Wilkinson, 2005). They are also present, to some degree, in the rhetoric of recent health policy concerning CHD in the UK (see Department of Health, 2000).

#### *The cholesterol hypothesis*

Aronowitz (1998) suggests that by the early 1960s both biomedical professionals and the lay public had accepted the idea that the risk of CHD was linked to eating high fat foods and hypercholesterolaemia (high blood cholesterol levels). Yet the cholesterol hypothesis has been highly contested at various points in its history. Garrety (1997) shows how 'the facts' about dietary fat and cholesterol and CHD became established despite ongoing controversy about the supporting evidence. The process by which these facts emerged involved the work and interaction of a variety of actors. Garrety argues, for example, that messages about dietary cholesterol were enthusiastically received by the lay public because this was in keeping with lay health movements' focus on diet, and that the food industry also drew on the hypothesis to develop and market 'healthy' products such as polyunsaturated margarine. Findings announced in 1984 as the first conclusive evidence that lowering cholesterol reduces heart attacks were, in fact, those of a clinical trial that targeted men with particularly high levels of cholesterol using a

cholesterol-lowering drug. This nevertheless led to national recommendations about dietary fat and cholesterol intake for everyone, defining a large proportion of the population to be at risk. Garrety (ibid: 753) concludes that by the time the results were announced:

‘the cholesterol hypothesis had been inside the boundary of orthodox medical knowledge for a quarter of a century. Most people were unaware, or did not care, that the ‘definitive proof’ was still lacking. The scientific ‘evidence’ had simply become irrelevant’

Aronowitz (1998) argues that hypercholesterolaemia was transformed during the 1980s from being a lifestyle issue of marginal interest to clinicians, to become fully established as a CHD risk factor to be seen as a medical problem to be managed in the clinical setting. He attributes this to the introduction of drug therapy and the creation of national recommendations on cut-off points for cholesterol levels. Writing from his north American perspective, he argues that the incentive for doctors was the creation of a reimbursable medical diagnosis with a specific definition and treatment. More recent controversy flared up in the late 1980s and early 1990s, when biomedical papers were published that again questioned the link between blood cholesterol levels and CHD and suggested that there was a link between low blood cholesterol and increased mortality from other causes (see Petersen & Lupton, 1996, pages 44-45 for details).

### *Public Health Discourses*

Health promotion discourses concerning CHD have been criticized for focussing on lifestyle to the exclusion of other possible risk-factors, and for oversimplifying epidemiological evidence, for example associating lifestyle modifications with avoiding disease rather than decreasing risk of illness

(Davison et al., 1992; Hunt & Emslie, 2001; Petersen & Lupton, 1996). Brett (1991) argued that professional, media and food industry presentations of cholesterol foster false expectations about its management and is particularly critical of the message that ‘serum cholesterol is universally lowered by prudent dietary habits’ (ibid: 645). Davison et al (1992: 677-8) argue that: ‘health educators and product marketing professionals have waged an intense public campaign to place lifestyle at the centre of CHD causation’. The implication within this discourse is that heart disease is largely attributed to either ignorance or lack of self-discipline, leaving little room for any uncertainty about the controllability of the disease (Davison et al., 1991, 1992). Davison et al (1989, 1991, 1992) argue that this construction of CHD is imbued with moral positioning. Sachs (1996: 637) has similarly described hypercholesterolaemia as: ‘a condition redolent with blame for unhealthy living’.

These observations about public health campaigns concerning CHD and cholesterol exemplify a wider critique of public health discourses, which emphasise personal responsibility, premised on the notion that illness can be avoided through identifying and acting on risk factors (Petersen & Lupton, 1996). Drawing on Greco (1993), Lupton (1995: 90) argues that:

‘failure adequately to control risk through strength of will has become a form of irrationality, or evidence of the inability to master the self. Disease has become linked to the state of one’s moral qualities rather than individual constitution’.

Death, therefore, becomes the ‘ultimate failure of self-control and rationality’ (Petersen & Lupton, 1996: 49). These arguments reiterate points made in the

first part of this literature review concerning the expression of contemporary rationales of risk and individual agency within the field of health.

### *Genetic models*

So far this section has argued that a risk factor model for CHD emerged in the second half of the twentieth century, bringing with it the idea that CHD was essentially predictable and preventable. This model places great emphasis on individual behaviour, framed as 'lifestyle' factors such as diet and exercise, and on measurable physiological factors such as blood pressure and blood cholesterol levels. Socio-historical accounts of CHD have, to date, made very little reference to specific genetic models, other than general references to family history. Davison et al (1989) writing in an era that predates the completion of the Human Genome Project, argued that there were different levels of interest in heredity and CHD in different branches of medicine. They suggest that there was some interest in epidemiological literature in genetic factors, which were thought to account for a moderate component of the variation in cholesterol levels, blood pressure and physical fitness. They argue that within clinical medicine interest lay mostly in the familial hyperlipidaemias and that materials for primary health care and health education paid relatively little attention to familial risks.

The first part of this review suggested that a large number of social analysts believe that common conditions are being reframed in genetic terms. The following discussion is intended to show the origins of these ideas. While there has been longstanding interest in the construction of genetic disease (see

for example Yoxen, 1982), in the field of the sociology of science and technology there has been growing interest in the development of molecular genetics and the re-construction of what counts as genetic disease. Martin (1999: 518), for example, suggests that a more ubiquitous genetic model of disease is emerging in biomedical discourses:

‘there has been a shift from an account of disease based on ‘classical’ genetics and the inheritance of deleterious genes, to one which explains many common acquired pathologies in terms of errors in the way gene (sic) are regulated’.

This focus on errors in the expression and regulation of ‘normal’ genes forms part of a wider shift toward describing pathology at the molecular level.

Martin and Kaye (1999) argue that this has resulted in a number of conditions being reclassified into subcategories on the basis of their molecular biology e.g. breast cancer, asthma and diabetes. Further, many common diseases are now thought to incorporate a ‘genetic’ subset in which there is a clear association between certain variants or mutations and the occurrence of disease e.g. BRCA genes and breast/ovarian cancer. Therefore, a new model of genetic disease has emerged that encompasses both explanations in terms of gene function at the molecular level for the underlying pathology, and the idea of genetic predispositions for common acquired conditions. It is notable that describing pathology in terms of gene function can apply to all cases of disease and does not relate to inherited differences between people, whereas the idea of genetic susceptibility depends on the inheritance of specific genetic mutations or variants.

A vision of a genetic future for understanding, managing and treating common conditions emerged in the 1990s. In this vision, common conditions are

constructed as polygenic (i.e. involving many genes), multifactorial conditions, in which susceptibility is conferred by the interaction of numerous susceptibility or complex disease genes; genetic research will lead to new classification systems, aiding diagnosis, and to the development of susceptibility/predictive testing, new treatments and targeted treatments (see for example Bell, 1998; Cm 5791 - II, 2003; Department of Health, 1995; Mathew, 2001; McCarthy, 2000). Matthew (2001: 1013) epitomises this view:

‘If the promise of the genome sequence is even partially fulfilled, the next decade will see genetics spreading rapidly beyond the confines of specialist centres to impact on the diagnosis and management of common disorders in primary care’

CHD features as a central example in this vision. In the UK, for example, it is one of the conditions that form the focus of the genetics White Paper (Cm 5791 - II, 2003). This offers a vision of predictive genetic risk assessment for CHD, accompanied by personalised lifestyle advice and personalised therapeutic drugs. CHD is certainly the focus of research activities for firms engaged in work with large biological sample collections (Martin & Kaye, 2000). Indeed, it is one of the key conditions cited as a focus for the nationally-sponsored UK Biobank (The UK Biobank, 2004; The Wellcome Trust, 2003). As has already been noted, commentaries on the potential social and cultural impact of developments in the genetics of common conditions also cite CHD as a key example (see for example Davison, 1996; Davison et al., 1994; Lippman, 1998). The idea of a genetic future for coronary heart disease is firmly embedded within (some) biomedical, social science and policy discourses. Nevertheless, there has not been a detailed analysis of the recent constructions of CHD that elucidates how this picture is emerging, or how widely these genetic models have currency.



### *Summary*

This section has argued that a ‘risk factor’ model of CHD emerged in the second half of the twentieth century, which also saw the reframing of CHD from an intractable degenerative disease of ageing to a condition that is largely predictable and preventable. This time period also saw the establishment of the cholesterol hypothesis, although controversies have continued to flare up around this. The ‘risk factor’ approach to prevention of CHD has been criticised for privileging individual level factors, positioning individuals as responsible for avoiding illness through adherence to appropriate ‘lifestyles’. It has been argued that a genetic model for common conditions, including CHD, emerged in the 1990s, accompanied by a rhetoric concerning predictive or susceptibility testing and personalised preventative strategies for such conditions. This has been noted and discussed widely in social analyses of genetic developments. It is notable, however, that although socio-historical accounts of CHD make reference to family history of CHD as a risk factor, detailed studies exploring the emergence and wider acceptance of a genetic model of CHD are absent.

### **Lay constructions**

#### *Lay constructions of CHD*

Studies within general populations about the causes of heart disease have found that family health histories and personal actions and attributes all figure large. Davison et al. (1991:5) have argued that the idea of the ‘coronary candidate’ or ‘the kind of person who gets heart trouble’ is central to lay accounts of CHD.

They describe three main elements that contribute to coronary candidacy: physical appearance, social and personal information: Physical aspects include obesity, in particular, and evidence of lack of fitness. Important aspects of social information are a family history of heart trouble, occupational factors (such as mental or physical stress, physical inactivity, and poor work environment), and geographical location (relating to ideas about local habits and economic and environmental influences). Personal information relates to individual behaviour and disposition. Smoking, eating large quantities, particularly of fatty foods, and high alcohol consumption were strongly associated with candidacy, as was a tendency to be nervous, worry a lot, or anger easily. The model clearly includes both 'lifestyle' factors and areas not perceived to be within the control of the individual, such as heredity, upbringing, relative wealth, occupational risks, climate or pollution. Emslie et al. (2001) re-evaluate the role that gender plays in ideas about heart problems and coronary candidacy. They note that when respondents in their study cited specific examples of the 'coronary candidate' or of 'the last person you'd expect to have heart trouble', these were both almost without exception male. In other words, men are perceived as both the most likely and the least likely to suffer heart disease in lay constructions, leading Emslie et al (ibid: 203) to suggest that women are 'invisible' in lay accounts.

Davison et al. (1992: 683) argue that, because lay models of CHD recognise that health is related to heredity, social conditions and the environment, as well as personal behaviours, they may be 'more in step with scientific epidemiology than the lifestyle-centred orientation of the health promotion world'. Davison

and colleagues (Davison et al., 1991: 14) argue that a striking feature of coronary candidacy is a recognition that it is a 'fallible system', in other words many people who do not fit the candidacy profile will become ill or die of CHD ('anomalous deaths') and not all identified candidates succumb to CHD ('unwarranted survivals') (ibid:16). Thus the notion of candidacy is concerned with increased risk - a random element remains.

Lupton and Chapman's (1995) study of lay peoples' responses to the cholesterol controversies of the early 1990s suggests that lay constructions of high cholesterol fit neatly within the notion of coronary candidate. This suggests that raised cholesterol is associated with strongly moralistic overtones, linking the condition with bodyweight and diet. Yet, complexity and uncertainty associated with cholesterol was also acknowledged. This included, for example, references to a propensity for a particular metabolism, or for raised cholesterol. This meant that raised blood cholesterol was not necessarily related to appearance and lifestyle, but was also down to luck or heredity:

'It was commonly noted that some individuals, regardless of their ingestion of dietary cholesterol or fats, simply manufacture high levels of cholesterol, while others may have low blood cholesterol despite their 'unhealthy' diet (ibid: 489).

#### *Lay constructions of a family history of CHD*

Davison et al (1989) have argued that in lay constructions, an hereditary input to CHD operates in a number of ways. One can inherit discrete physical attributes, such as high blood pressure or 'heart attacks', one's constitution e.g. a weak heart, a tendency to be fat, or elements of personality and behaviour such as being a worrier or being laid-back. They argue that inheritance is a

subject 'shot through with a strong flavour of chance' (ibid: 335) and because of the complexity of heredity and the importance of other causes, it was used more to explain existent cases of heart trouble than to predict cases.

Ideas about a hereditary element to CHD are widespread. Hunt et al (2000a) found, for example, that a fifth of their respondents thought they had a family history of heart disease or heart trouble. Perceptions of a family history of CHD are patterned by gender, class and age (Hunt et al., 2000b, 2001; Watt et al., 2000). Overall, older people were less likely to perceive this family history of CHD. Men seemed to need more affected relatives than women in order to assess themselves as having a family history of CHD. Working class men and women tended to attribute deaths to old-age for relatives at younger ages than did middle class people. Overall, working class men were particularly ambivalent about whether they had a family history of CHD.

### *Health Responsibilities*

The findings of studies concerning lay constructions of CHD fit with other studies of lay constructions of health and illness. In summarising such studies, Blaxter (1997: 750) comments that lay models of the causes of illness cannot be simply described, arguing that lay respondents tend to 'move back and forwards between concepts of cause which seem opposed', such as responsibility for health-related behaviour, having the right mental attitude and ideas about chance, luck and inevitability. Blaxter considers whether notions concerning health inequalities, present in the epidemiological literature discussed in the preceding section, are evident in lay accounts. Observing that

that working class people, in particular, are likely to reject the idea of inequalities in health, she draws on Crawford (1980), to suggest that this is tied up with a 'healthist' culture that links illness to moral failings. She argues that it is not surprising that the idea of 'not giving in to illness' is prominent in working class respondents accounts, suggesting that this 'can be seen as a claim to moral equality even in the face of clear economic inequality' (Blaxter, 1997: 754). Such studies show that health and illness are tied to personal responsibility as much in lay accounts as in professional constructions.

Although some have suggested that providing information about genetic susceptibility to disease may lead to greater fatalism (see for example Senior et al., 1999, 2000), others, such as Hallowell (1999), Novas & Rose (2000) and Polzer et al. (2002) argue that, within a logic of risk management and individual responsibility, such information opens up new areas of responsibility. The first part of this chapter discussed Novas & Rose (2000) notion of 'genetic prudence', which involves the duty to become informed of future health risks based on genetic information and to undertake risk management in relation to oneself and those to whom one is connected. Specific studies concerning genetic responsibility will be discussed shortly, but at this point the notion can be related to lay accounts of CHD.

Davison et al. (1992) argued that in popular explanations of CHD, non-control areas, particularly the field of luck/fate, are subsidiary to lifestyle and have suggested that the scope to change individual behaviour was seen as 'counter-acting' the effects of heredity' (Davison et al., 1989: 338). Hunt et al (2000a)

report that people who assess themselves to have a family history of CHD attribute even greater importance to lifestyle and were no more or less fatalistic than other respondents overall. Ponder et al (1996) report that nearly half of participants with affected relatives did not think this was relevant to their own risk of CHD. Personal risk was assessed through interpretations of family experiences and events which tended to lessen the impact of inherited risk in a number of ways: the illness could be seen as particular to that relative due to lifestyle, environment or chance; the inherited susceptibility could be counterbalanced by the participant's own actions; or the risk was lessened because the participant 'did not take after that side of the family' (ibid: 489).

These studies suggest that the notion of genetic prudence or responsibility is compatible with lay accounts of CHD and that a commitment to individual responsibility for CHD is not disrupted by its hereditary aspects. The last reference to reduced risk through not 'taking after that side of the family' also suggests that the notion of genetic responsibility must take account of lay constructions of heredity, since the importance of genetic risk factors for self and others may depend to some extent on how these risks are understood. Although there have been a number of studies of lay models of heredity (see Emslie et al., 2003 and Richards, 1996a, b, 1997), the implications of these models have not been discussed in relation to genetic responsibility.

#### *Lay constructions of familial hypercholesterolaemia*

There has been relatively little research on patients' experiences of FH, specifically, and existing work is mainly concerned with the psychological

aspects of the diagnosis (reviewed in Marks et al., 2000). Lambert & Rose (1996) provide the only broadly sociological analysis of FH. The study was based on interviews with people with a variety of familial hyperlipidaemias (FH in the main). Lambert & Rose (1996) argue that FH is defined by abstract information unrelated to feelings of well-being and that its treatment involves action on the body although the results of these actions cannot be directly perceived by patients. The central concern of this study was to explore how people make sense of the 'disembodied knowledge' concerning FH, in other words, the interplay between this knowledge and the patients' embodied understandings.

Although this is not the main focus of their study, they do provide some evidence about how patients construct the inherited aspects of the condition, finding that prior to diagnosis, most interviewees had not been aware that high cholesterol could be associated with inherited factors. Indeed they generally became aware of the significance of FH in terms of their family history in retrospect i.e. only after their own diagnosis of high cholesterol or CHD. Lambert & Rose (1996) touch briefly on ideas about the significance of FH to patients' life plans, finding that the condition is not deemed relevant because high cholesterol was seen as an 'everyday thing' (ibid: 79). This suggests that the idea of genetic prudence may have less resonance in this case. Lambert & Rose (1996: 79) conclude that:

[participants'] comments about the implications of this hereditary disorder integrate it effectively into a view of human life that acknowledges individual variation, the multifactorial causation of ill health, and the normality of human imperfection'.

However, there is some evidence that patients may make a distinction between those with FH and people with raised cholesterol for other reasons, perceiving people with FH to be less culpable for their status (see Senior et al., 2002; van Maarle et al., 2003)

### *Genetic responsibility*

As has already been argued, the possibility of providing people with information about genetic susceptibility to disease has been associated with two different responses: fatalism or genetic responsibility. Hallowell (1999) and Polzer et al. (2002) have explored lay constructions of genetic responsibility in the cases of hereditary breast/ovarian cancer (HBOC) and familial melanoma. Hallowell found that women at risk of HBOC felt an obligation to determine and manage their risks, inform others and encourage them to do the same. The study makes clear that public health discourses about genetic risk and responsibility also circulate among the lay people experiencing genetic technologies. According to the women interviewed, the motivation for gaining risk information and acting on this was largely concerned with other people's needs, including researchers'. Further, the women's responsibility was linked to current, past and future generations. Hallowell (1999: 616) comments that it is not clear the degree to which the sense of obligation felt by these women was connected specifically to genetic risks rather than health risks more generally: 'their accounts suggest that it was their acknowledgement of their social connections and associated obligations which led them to manage their own risk and inform others about their risks and encourage them to engage in risk management'. Hallowell questions whether men in similar



positions to women in her study would account for their behaviour in the same way. Richards (1996a: 258) has also commented on a possible gendered relationship to genetic risks and responsibilities, suggesting that ‘women usually act as genetic housekeepers for the kinship’.

This idea, however, is not supported by Polzer et al.’s (2002) study of genetic testing for familial melanoma risk, which involved both men and women. The study provides remarkably similar findings to Hallowell concerning people’s motivations for testing. Polzer et al. (2002) argue that participants felt a duty to know and manage genetic risks and to inform family members. As in Hallowell’s study, participants expressed a strong sense of obligation to raise the issues of genetic risk with family members not only to pass on genetic information, but also to encourage them to monitor themselves. While Polzer et al. suggest that responsibilities extend to blood relatives, Hallowell frames responsibilities in terms of social connections. Cox & McKellin’s (1999) work on how families with Huntington’s disease (HD) construct risk suggests that even among blood relatives, ideas about genetic closeness are shaped by social connections. They comment:

‘Factors such as geographic and social proximity to an affected family member are as important as biological ties in explaining test candidates and their families’ intersubjective constructions of hereditary risk’.  
(ibid, 1999: 641)

This reinforces the argument that the notion of genetic responsibility must be considered alongside lay constructions of heredity.

These studies of genetic responsibility and other studies concerning the communication of genetic information within families, for example d'Agincourt-Canning (2001), Forrest et al. (2003), Green et al. (1997) and Kenen et al. (2004) have been undertaken with samples drawn from clinical genetics (and relate almost exclusively to HBOC). Furthermore, the rationale for obtaining genetic information in these cases has related to making behavioural changes, and self and medical surveillance. Novas & Rose's (2000) discussion of genetic prudence draws on the example of Huntington's Disease. Cases such as FH and CHD which are managed outside of clinical genetics and for which prophylactic therapies are available have not been considered.

### *Summary*

This section has suggested that lay constructions of CHD, embodied by the notion of the 'coronary candidate' involve a range of factors including 'lifestyle' and heredity. Studies of lay constructions of CHD or a family history of CHD suggest that lifestyle is seen as having the potential to counteract hereditary susceptibility. These constructions are congruent with wider studies concerning lay accounts of health and illness in which personal responsibility represents a dominant theme. The review has suggested that there is a paucity of studies relating specifically to patients' experiences of genetic susceptibility to CHD or other conditions for which prophylactic therapies are available. Existing studies concerning the provision of predictive genetic information suggest that acquiring such information is associated with a sense of genetic responsibility relating to oneself and to others.

## **Patients' associations and the production of knowledge**

The inclusion in this study of HEART UK, the health charity originally formed as a patients' association to support people with hereditary lipid disorders, was based on recent sociological discussions about the role of such groups in the production of knowledge. These discussions are linked to the increasing acceptance in sociological debates of the notion of lay knowledge, and the positioning of lay health groups as important sites where expertise is renegotiated and lay knowledge expressed. This section briefly reviews the discussions about lay expertise and then turns to the literature on lay health groups and their role in the production of knowledge.

### *Lay expertise*

In the previous sections, the literature on disease constructions was presented under two distinct headings, biomedical constructions and lay constructions, reflecting the way research on these topics is often organised. Nevertheless, these two categories create an artificial divide to some extent. As Davison et al. (1989: 329) have argued, medical and popular concepts of health and illness 'merge almost imperceptibly' in contemporary Britain, calling into question the idea that they form two distinct types of accounts:

'The non-professional majority are habitual users of the medical idiom when illness is under discussion, and the professional minority remain members of the wider society, sharing the common framework of cultural and moral norms.'

Shaw (2002: 287) similarly asks 'how lay are lay beliefs?', arguing that the expert or biomedical model is integral to contemporary 'common sense' understandings of health. He argues that patients, particularly with chronic

illness, become expert in the biomedical knowledge associated with their condition:

‘but also utilizing their experience of suffering as a way of negotiating or critiquing that knowledge...this expertise can challenge the doctors’, particularly the non-specialists, authority’  
(ibid: 295).

These comments exemplify an increasing recognition of the notion of lay or experiential knowledge in sociological writing.

Discussion of lay knowledge has been integral to critiques of the production of scientific/medical knowledge and of the development of science/health policy (see for example Kerr et al., 1998; Popay & Williams, 1996; Williams & Popay, 1994; Williams et al., 1995; Wynne, 1996a, b; Yearley, 2000). These have contrasted expert claims, based on ‘universalistic knowledge’ (Yearley, 2000) against lay people’s ‘situated understandings’ (Lambert & Rose, 1996: 80) based on experience and knowledge of local conditions. This critique has both epistemological and political elements and is enrolled in calling for the involvement of lay people in the production of scientific knowledge and in science/health policy. The idea of lay knowledge or expertise is now relatively established in social science literature (although see Prior, 2003). Recent debate has concerned its definition, extent and role (Collins & Evans, 2002; Jasanoff, 2003; Rip, 2003; Wynne, 2003). The notion is also slowly gaining recognition in some policy circles in the UK. It is recognised, for example, in the Department of Health’s *Expert Patient* initiative (Department of Health, 2001a) and through the Department’s involvement agenda (see Baggott et al., 2004, page 318 for details).

### *Lay health groups and knowledge production*

Lay health groups have been prominent in the analysis of lay knowledge and expertise and its relationship to biomedical science and policy. Such groups offer a potentially important site of lay knowledge and a point of contact or negotiation between lay people/patients and clinicians and researchers, at a collective or institutional rather than individual level. There seems to be agreement that there has been a growth in the numbers of lay health groups from the 1970s onwards (Allsop et al., 2004; Epstein, 1995; Kelleher, 1994; Rose & Novas, 2004; Wood, 2000). While the literature on lay health groups is not new (see for example Robinson & Henry, 1977), there is also agreement that lay health groups have been relatively little studied within the social sciences (Epstein, 1995; Rabeharisoa & Callon, 2002; Wood, 2000).

Lay health groups can mean a great many things including self-help groups, patient organisations, advocacy or activist groups and medical research charities. Health professionals can be involved in these groups in various ways, e.g. as founders, members or staff. This raises the question as to what counts as a lay health group (or perhaps to borrow from Shaw, 2002, how lay are lay health groups?). Wood's study of disease-related patients' associations attests to the difficulty of delimiting these groups, with Wood admitting to having to apply pragmatic inclusion criteria as to whether an 'association seem[ed] to be patient-led and independent' (Wood, 2000:23). These groups may engage in a variety of 'inner-focused' activities such as self-help or support groups and 'outer focused' activities such as educating professionals and lobbying for resources or recognition (Kelleher, 1994). The extent to

which these different activities might be considered a challenge to medical authority has been a consistent theme within the literature on lay health groups. It could be argued that 'outer focused' activities present more potential for overt challenges to biomedical expertise or for renegotiating the status of lay knowledge. Kelleher (1994: 111) argues that while activities such as support groups could be seen as complementary to medical work, they provide an implicit challenge to biomedical knowledge, displaying 'a subversive readiness to question the knowledge of doctors and to assert that experiential knowledge has value'. Rose & Novas (2004) similarly argue that such groups offer a space where biomedical science can be problematised.

There are a small number of documented cases in which lay health organisations have attempted to influence knowledge production. Brown (1995b: 93) suggests three sets of aims for lay health advocacy groups, which could be usefully extended for thinking about lay health groups more widely:

1. To increase resources for the prevention and treatment of already recognised diseases
2. To gain increased recognition of unrecognised or under-recognised diseases
3. To establish knowledge about aetiological factors in recognised diseases.

The aims of the AIDS activists in bringing about changes to access to clinical trials could be placed within the first category (Epstein, 1995) (this will be discussed in more detail shortly). Within the second category, lay health

groups have been involved in successfully establishing, or trying to establish, a number of important diagnostic categories, including sudden infant death syndrome, post traumatic stress disorder, Alzheimer's Disease and repetitive strain injury (see Arksey, 1994; Fox, 1989; Johnson & Hufbauer, 1982; Scott, 1990). Brown's own work (Brown, 1992, 1995b) on toxic waste and popular epidemiology relating to a cluster of childhood leukaemia in Woburn Massachusetts provides an example of the third category. McLean (1990) provides a further example concerning the influence of the National Alliance for the Mentally Ill, in shifting ideas about the aetiology of schizophrenia from family-based explanations to biological or organic models.

In these studies, lay health groups successfully employed a range of methods including lobbying state funding bodies and politicians to gain research funding, involvement in official policy bodies, cooperating with and participating in professional organisations, disseminating information to health care professionals, organising conferences to bring interested parties together, public awareness campaigns, initiating and funding research, helping to provide research subjects or materials and even involvement in undertaking the research.

#### *Lay health groups influencing research practices*

Recent discussions have highlighted cases where lay health organisations have influenced the very practice of research. One of the most well known of these is Epstein's work on the influence of AIDS activists. Epstein (1995: 409) demonstrates how these activists became:

‘genuine participants in the construction of scientific knowledge...[effecting] changes both in the epistemic practices of biomedical research and in the therapeutic techniques of medical care’.

A major issue was the design of randomised controlled clinical trials concerning therapies. From the activists’ perspective, entry criteria aimed at providing ‘clean data’ failed to take account of the social realities of the time. Activists recognised that clinical trials provided a way to access potentially useful therapies that may otherwise have been unobtainable. Clinical trials were seen, therefore, as both scientific experiment and contributing to health care, and equity of access was a major issue. Activists were also aware of arguments within biomedical science about the design of clinical trials, concerning the degree to which trial populations should be homogeneous or reflect the heterogeneity of intended target populations. Epstein argues that by bringing together methodological, epistemological, moral and political arguments the activists successfully:

‘won support for a number of modifications in trial design including the use of broader entry criteria, more diverse subject populations, and concomitant medication’ (ibid: 424).

The case of the Association Française Contre les Myopathies (AFM), the French muscular dystrophy association (Callon & Rabeharisoa, 2003; Rabeharisoa, 2003; Rabeharisoa & Callon, 2002; Rabinow, 1999) has received considerable recent attention. Callon & Rabeharisoa (2003) discuss how the AFM set about gathering information about the condition in the form of photos, films, written accounts and conducting surveys. These methods were used as a means of formalising and publicising members’ knowledge about the condition, which led to collaborative research with biomedical scientists and clinicians. This resulted in, for example, the identification of different forms of



the disease and clarification of clinical profiles. Rabinow (1999) argues that AFM's collaboration with CEPH, France's most prestigious genomics laboratory, not only managed to get the muscular dystrophies onto the research agenda, but also influenced research practice, particularly the wide and rapid public dissemination of findings. It created a research model both more 'entrepreneurial' and more 'civic' than state-funded laboratories could provide (ibid: 44). Heath et al. (2004), Rapp et al. (2001), Rose & Novas (2004) and von Gizycki (1987) provide further examples of collaborative work and innovative research relationships in which patients' organisations have influenced or controlled how knowledge is produced and used.

On the basis of these examples, Rabeharisoa and Callon (Rabeharisoa, 2003; Rabeharisoa & Callon, 2002) propose that there are now three different models concerning the engagement of lay health organisations with research, the auxiliary, emancipatory and partnership models. Rabeharisoa (2003) argues that the auxiliary association either delegates decisions about research priorities and knowledge dissemination to the biomedical specialists or acquires biomedical expertise in order to enter into the discussions. In the emancipatory model, professional constructions may be challenged or rejected as groups strive to assert their own collective identity, as exemplified by some disability organisations. It is suggested that cases like the AFM represent a third model, the partnership model. This has two main characteristics, that the organisation retains control of their research policy, and that 'patients are specialists' partners in their own right' (ibid: 2131). Patients' experiences are prioritised

and the role of the organisation is to collate this expertise and communicate it to the biomedical professionals in a way they can understand:

‘if patients are to be regarded as “experts in experience”, their knowledge of the disease must be formalised in such a way as to demonstrate its value’  
(Rabeharisoa & Callon, 2002: 62).

The organisation, therefore, has a unique role as custodian and translator of collective experiential knowledge.

Both Rabeharisoa & Callon (Rabeharisoa, 2003; Rabeharisoa & Callon, 2002) and von Gizycki (1987) point out that the potential for any patient organisation to influence knowledge production depends to an extent on the existing players and state of knowledge in their field, including the statutory authorities, research institutions, other voluntary organisations and industrial interests. A collaborative or partnership model may be more likely to emerge where the research environment is unstructured and specialist scientific or medical groupings are absent.

While the partnership model relates specifically to the contribution of collated and formalised experiential knowledge to research, it is likely that many patients’ associations would recognise a role in drawing together the views and experiences of patients in order to influence the direction of funding, research and medical practice. This may involve analysing contacts with helplines, surveying members and drawing on the discussions of local meetings.(Allsop et al., 2004; Baggott et al., 2004).

So far this section has argued that studies of lay health groups have contributed to a wider reappraisal of expert knowledge. They are seen by some analysts as part of a broader social movement in which people affected by biomedical science want to have a say in its making. Epstein (1995: 428), for example argues:

‘To varying extents, these groups challenge the hierarchical relations between laypeople and insist on the rights of those affected by biomedical science to participate in its production’.

Rabeharisoa & Callon (2002) similarly argue that the involvement of patients’ associations in research is part of a broader trend which has seen the reshaping of relations between experts and the groups who are likely to benefit from their expert knowledge. It has also been argued that these groups have the potential to change the way science is conducted.

### *Dissenting voices*

Stockdale’s (1999) study of the Cystic Fibrosis Foundation (CFF) in the US challenges these arguments about patient organisations as a new democratic form associated with new forms of collaboration in the production of knowledge. He argues that the decision of the CFF to focus its considerable research funds to finding a ‘cure’ for cystic fibrosis, concentrating on gene therapies, was not necessarily widely supported by people with CF and led to other activities being marginalised. Similar tensions about priorities were noted in a number of the case studies already discussed. Stockdale also reports that, in the case of the CFF, there were poor relations between researchers and the researched, and that the CFF actively excluded patient participation in

discussions about clinical practice and research. Stockdale (1999: 585)

comments that

‘the experience and relationships in the CF case are a striking example of the lack of consumer involvement in the research process’.

Williams’ (1989) study of the National Ankylosing Spondylitis Society (NASS) provides a second example where lay knowledge did not seem to be prominent in the culture of the organisation. At the national level, there was a greater focus on self-management rather than any kind of collective or interpersonal activities, which ‘seems to have been sustained by the dominance of middle-class professional values and interests’ (Williams, 1989: 152).

However, this ideology was not necessarily reproduced by lay members at the local level. The findings of these two studies suggest that research on patients’ organisations may need to pay attention to how widely patients’ knowledge is recognised and is contributing to knowledge production.

#### *Patients’ associations as biosociality*

Rose and Novas (Novas & Rose, 2000; Rose, N., 2001; Rose & Novas, 2004) link ideas such as those discussed here concerning changing forms of expertise and the growth of patients’ associations to their critique of geneticisation, discussed in the first part of this literature review. To recap, they argue that genetic information is collectivising as well as individualising, and it is collectivising partly through biosociality. In other words, people are increasingly joining into groups based on shared biological identities and that such groups are:

‘not merely demanding public provision and rights, but making their own claims on the deployment of biomedical technologies and the direction of biomedical research’  
(Rose, N., 2001: 19).

Indeed, they argue that, in some cases, active membership of a patient organisation is reconfigured as a duty. Rose & Novas (2004: 451) argue that the biosocial communities represent a new kind of active biomedical citizenship and that:

‘in a certain political, cultural and moral milieu, this idea of activism in relation to one’s biomedical condition becomes a norm. Activism and responsibility have now become not desirable but virtually obligatory’.

Taking the case of Huntington’s disease, Novas & Rose (2000: 506) argue that ‘the responsible-genetic subject becomes active in the shaping of the enterprise of science’ through engaging in discussion, donating money, fundraising and participating in research.

Biosociality should not necessarily be seen as a widespread practice. As Callon & Rabeharisoa (2004) report in their work on the muscular dystrophies, there are people who resolutely refuse a genetic identity and a genetically-identified collective. In general, Rapp et al. (2001) argue that certain kinds of people are more likely to engage in biosociality than others. Commenting here about genetic identity, they argue:

‘the class-inflected etiquette of voluntary organizations may feel more comfortable for middle-class families who are used to assuming “new” professional identities and seeking help from specialized sources. Working-class, and especially racial-ethnically marked populations ... may find alternative sources of support that do not focus on health problems or genetic identity categories’  
(ibid: 397).

There may also be attributes of particular conditions that are linked to the likelihood of them becoming part of identity practices. For example, Allsop et

al. (2004) suggest that in UK and in the US there are fewer high profile groups formed by patients or carers in the area of heart and circulatory disease compared with other disease areas studied. They comment (ibid: 744) that: ‘[it] does not appear to arouse feeling of anger or resentment, or pose a threat to identity’ in the same ways as other areas included in their study.

### *Summary*

This section has argued that the boundary between lay and expert has increasingly been problematised in sociological discussions and that studies of lay health groups have played a prominent role in this. A number of analysts have argued that such groups have the potential to influence the production of biomedical knowledge through contributing to or taking charge of research processes, based on the experiential knowledge and priorities of its members. The extent of the emergence of such novel relationships regarding expertise within patients associations is not clear and the idea that lay health groups privilege lay knowledge through their discourses and activities has also been contested.

### **Disease Constructions: Summary**

This review has suggested that both biomedical and lay models of CHD are dominated by a focus on ‘lifestyle’ which strongly links CHD prevention to personal responsibility. While there is a clear role for heredity in lay models, this is seen as secondary to lifestyle. This is consistent with a genetic model that recently emerged in biomedical discourses, which is premised on the identification of genetic susceptibility to CHD and provision of personalised

preventative regimes. This review has argued that, although references to a genetic model for common conditions such as CHD can be widely found in the social analyses of genetic developments, the degree to which such models have actually become established in biomedical discourse has yet to be studied. It has also been suggested that existing studies concerning lay constructions of genetic risks and responsibilities have been undertaken mainly within genetic clinics and relating to conditions for which prophylactic therapies are absent. These observations about the current state of scholarship in these areas reinforce those made earlier about the state of empirical work regarding geneticisation. In short, with the exception of hereditary breast/ovarian cancer, there have been few studies that relate to the genetic construction of common conditions. This thesis, which looks at both biomedical and lay constructions of CHD and FH, is therefore novel in as much as (1) it is a study of genetic susceptibility for a common condition for which prophylactic therapies are available and (2) it focuses on a common condition for which there are already strongly established biomedical and lay models. Finally, the review suggests that lay health groups are seen by some analysts as sites where biomedical knowledge is produced, or at least where lay or experiential knowledge is collated. This suggests that such groups might be important places to study disease constructions. The thesis adds to the limited body of research concerning patients' associations and the construction of knowledge.

## CHAPTER 3: RESEARCH DESIGN AND METHODS

### 3.1 INTRODUCTION

This chapter explains the rationale for the research design and how this developed, and provides a detailed account of the research process. This study employed a number of qualitative methods drawing upon several different types of data sources. The main methods were:

1. **Analysis of biomedical literature**, including a small number of selected recent commentary papers on CHD, and publications of HEART UK professional members and of the Simon Broome Register Group.
2. **Ethnographic work with HEART UK**, involving observation of the organisation's public activities and analysis of the documents it produces.
3. **Interviews**, with senior members of HEART UK, and with patients with FH recruited through a lipid clinic.

Table 3.1 provides a summary of how these different data sources relate to each research question. To recap, the main questions ask how FH and CHD are constructed in recent biomedical literature, and how HEART UK and patients with FH construct these conditions. These questions are intended to incorporate exploration of both explicit accounts of the aetiology of CHD and FH and the constructions of these conditions that are embodied in HEART UK's activities and in FH patients' talk about their experiences and actions as a result of diagnosis.



**Table 3.1: Sources of research data for each question**

Research Questions	Sources of data
1. How are FH and CHD constructed in the recent biomedical literature?	Analysis of : <ul style="list-style-type: none"> <li>• a small number of commentary papers on CHD</li> <li>• selected publications of HEART UK professional members</li> <li>• Simon Broome Register group publications</li> </ul>
2. How does HEART UK construct FH and CHD?	<ul style="list-style-type: none"> <li>• Analysis of written materials concerning the aetiology and treatment of FH and CHD</li> <li>• Analysis of written materials concerning the aims, objectives and activities of the organisation including collaborative work, and the research, technologies and policies supported.</li> <li>• Observation of public events and other activities.</li> <li>• Interviews with staff, trustees and senior members of the organisation.</li> </ul>
3. How do patients with FH construct FH and CHD?	<ul style="list-style-type: none"> <li>• Interviews with patients with FH</li> </ul>
4. What roles do patients play in the construction of discourses about FH and CHD within HEART UK?	<ul style="list-style-type: none"> <li>• Interviews with staff, trustees and senior members of the organisation.</li> <li>• Written information about the workings of the organisation and its history.</li> <li>• Observation of activities</li> </ul>

This chapter first discusses the rationale for selecting the case of FH and CHD. It then discusses the methods employed including discussion of how the research plan developed, sampling, access and recruitment, the conduct of the research and analysis. The last part of the chapter reflects on the research

process, considering ethical issues that arose in the research and reflecting on the limitations of the study.

### **3.2 CASE SELECTION**

The previous chapter suggested that the geneticisation thesis generated a great deal of discussion, but has been the direct subject of little detailed empirical research. The chapter also suggested that most empirical work concerning lay constructions of genetic disease had focussed on ‘classic’ Mendelian conditions, which tend to be relatively rare and untreatable, such as Huntington’s disease and Duchenne Muscular Dystrophy, or on hereditary breast/ovarian cancer (HBOC). While this provides an example of an hereditary risk of a common condition, it can be characterised as a condition that tends to involve radical and invasive treatments and a condition for which preventative therapies are absent.

The starting point for the research was to provide an empirical test of the claims of the geneticisation thesis. By focussing on common adult-onset diseases, moving away from a focus on conditions managed through obstetric and genetic services, the aim was to provide quite a stringent test of the limits of the thesis. Furthermore, this would provide an opportunity to compare whether the existing analyses of lay constructions of genetic disease had resonance in other settings. The task was therefore to identify a case where a genetic condition leads to risk of a common condition, where preventative therapies and treatments are available. The search was in some respects for a more ‘mundane’ condition. Bearing in mind suggestions in the literature that

lay constructions of genetic risks and responsibilities may be gendered (Hallowell, 1999; Richards, 1996) and the existing focus of work on HBOC, the aim was also to identify a condition that affects both men and women. The choice of FH/CHD was, therefore, a strategic decision, driven by the aim of testing theoretical concepts. This strategy can be seen as a form of ‘theoretical sampling’, understood as:

‘a process by which the researcher sets critical tests for the general validity of hypotheses and seeks to establish the conditions under which they do or do not hold’  
(Murphy & Dingwall, 2003: 113).

Drawing on the arguments outlined in the literature review about the lack of agency attributed to lay people within the geneticisation thesis, my original intention was to focus on lay constructions of FH and CHD, by studying patients with FH and the patients’ association for people with FH, the Family Heart Association. However, early in the study the FHA merged with the British Hyperlipidaemia Association (BHA), which was the national association for scientists and health professionals involved with lipid disorders, to form HEART UK. Since HEART UK aims to act as both a patients’ association and a professional body, this raises questions about the degree to which it can be thought to represent a site of lay discourses and actions. This necessitated some rethinking of the research strategy and questions. For this and other reasons which are fully explained in the following sections, by the end of the pre-fieldwork phase, the focus had broadened to include biomedical constructions of FH and CHD.

### **3.3 ANALYSIS OF BIOMEDICAL LITERATURE**

As discussed above and in the introductory chapter, it was not my original intention to undertake an analysis of biomedical constructions of CHD, but to draw on existing analyses against which my own work on patients' constructions could be set. My initial reading of the literature suggested that genetic models for CHD were emerging along with such models for other common conditions. However, I could not find any detailed analyses that focused specifically on recent constructions of the aetiology of CHD. In order to get a feel for the scope and nature of the field, I started to undertake searches of the biomedical literature concerning CHD and FH and accrued a large number of papers and chapters from biomedical journals and books concerning the causes and management of these conditions. Although initially I viewed this as secondary literature to be discussed in my literature review, I realised that this was actually primary data that should be analysed as such. At this point, I took the decision that the research should include some systematic analysis of the biomedical literature.

A full exploration of biomedical literature concerning CHD and FH would be beyond the scope of the thesis. Indeed, it could be the subject of one or more entire theses. There is a vast literature on CHD. For example, by January 2004 there were some 13,000 papers indexed in Medline under the MeSH heading 'coronary disease/etiology' and almost 2,200 under the heading 'coronary disease/genetics'. My initial plan was to analyse a small number of general accounts of CHD, and this analysis was undertaken early in the study. The merge between the FHA and BHA suggested the focus for further work, which

was undertaken later in the study. The thesis, therefore, came to focus on three specific areas of literature:

1. a small number of recent general accounts of CHD
2. papers by professional members of HEART UK
3. the publications of the Simon Broome Register Group.

The Simon Broome Register Group is a semi-autonomous research group concerning FH, which is loosely connected to HEART UK. The following sections will discuss the use of biomedical texts for studying disease constructions and the rationale for focussing on these three areas of literature.

### **Analysing biomedical texts**

There are potentially many ways of studying biomedical constructions of diseases including analysis of biomedical publications and undertaking interviews and ethnographic studies with biomedical professionals. This thesis mainly draws on biomedical publications, but also includes some interviews with clinicians and scientists, which are discussed in later sections of this chapter. Journal accounts of an area of science are highly accessible. They are the accounts most likely to be available to and seen by others (Hedgecoe, 2001a) and provide ‘potent markers of the state of knowledge in a particular field’ (Kerr, 2000: 854). The different methods for studying scientists’ accounts of a given field tend to elicit different sorts of discourses. One feature of journal accounts is that they tend to minimise ambiguity and controversy (Gilbert & Mulkay, 1994; Kerr, 2000; Kerr et al., 1997). Myers (1990a) argues that although arguments are not uncommon in science, they rarely surface in

the scientific literature. He proposes that the usual way of dismissing research in scientific papers is to ignore rather than to criticise it. Because differences and ambiguity are often hidden, it is useful to compare different texts as a way of highlighting the different ways of constructing particular ‘problems, ‘facts’ and artifacts’ (Kerr, 2000: 854). This requires sensitivity to the types of arguments and data that are included and those which are absent, and the kinds of understandings that remain implicit (Gilbert & Mulkay, 1994; Kerr, 2000). These principles guided my analysis of the biomedical literature concerning CHD and FH.

### **General accounts of CHD**

My initial analysis concerned general accounts of CHD within the biomedical literature, in order to gain an overview of this field. The aim of this analysis was to indicate what aetiological models of CHD are present in the literature, to show how a genetic model of CHD and alternative models of CHD are constructed, paying attention to the kinds of arguments and evidence that are enrolled and the aspects that are absent. This analysis identifies a number of different strands to aetiological models of CHD and types of arguments to support them, and this provides a framework for the subsequent analysis of biomedical constructions.

The analysis focused on commentary articles. This type of article includes reviews, editorials and discussion pieces. These discuss or draw together published research. They are distinguished from original research papers, which present new, previously unpublished research. The method of analysing

a limited number of commentary articles draws particularly on the work of Hedgecoe (2001a, 2002, 2003a) on the construction of genetic discourses in scientific review papers.

Review articles and other commentary pieces play an important and particular role in shaping scientific knowledge. Citing the work of Greg Myers (1990a, b, 1991), Hedgecoe (2001a: 878) suggests that:

‘review articles provide a textual space within which knowledge is constructed, allowing certain experimental reports to be seen as key papers. Even the idea of the discovery of a particular fact depends upon review articles to organize the claims and techniques in a particular direction’.

In other words, reviews shape scientific knowledge by selecting from existing research papers and putting these together to synthesise a particular narrative.

The review draws the reader:

‘into the writer’s view of what has happened, and by ordering the recent past, suggests what can be done next’  
(Myers, 1991:46).

They are, therefore, concerned not only with what has happened, but also with shaping the future. Hedgecoe (2003b) illustrates that commentary articles may well outnumber original research papers in emerging scientific fields. This volume of commentaries would not be warranted if they were only concerned with reporting on current knowledge, but can be understood in terms of their importance in constructing future visions. Following this logic, one would expect to find genetic discourses about CHD at least in commentary pieces, even if they were absent elsewhere.

This thesis analyses four commentary articles, selected from the fields of genetics, epidemiology and cardiology. The selection of these articles was pragmatic to an extent, in as much as they were chosen at the early stages of the study in order to help get to grips with the scope of arguments about CHD. An initial search of Medline was undertaken using the MeSH term ‘coronary disease’ and key word ‘causes’. This was limited to review articles in English from 1999-2003 and resulted in 79 papers. However, the majority were very specific, concerning interventions, therapies, specific risk factors, CHD in relation to particular syndromes or conditions, or specific demographic groups. The search yielded two general epidemiological articles concerning CHD risk factors and one general review concerning genetics and CHD. A second search of Medline using the key words ‘coronary heart disease’ and ‘genetics’ found 20 commentary articles in English published from 1999-2003, of which six provided general overviews, including the one mentioned above. The other articles concerned specific areas such as familial disorders, genetics and diet or lipid regulation. Further searches of Medline using other keywords and MeSH terms, eg ‘Etiology’, and the same limits as the searches above were abandoned because of the large datasets they produced. A search of the Eureka History of Science Technology and Medicine database was undertaken and this led to the identification of a further four biomedical overview papers published in English between 1999 and 2003. Taken together, these initial searches identified a dozen possible articles, from which four articles were selected for analysis. They were chosen because they offered relatively general and accessible accounts. They were also authored by eminent scientists in their



respective fields and were published in relatively influential (i.e. high-ranking) journals.

They are: **1.) Stephens and Humphries (2003) ‘The molecular genetics of cardiovascular disease: clinical implications’.** This review paper is specifically concerned with genetics and CHD. Professor Steve Humphries is one of the UK’s leading scientists in the field of cardiovascular (CV) genetics, Director of a British Heart Foundation funded centre for CV genetics and Chief Executive Officer of London IDEAS (London’s genetic knowledge park). Jeffrey Stephens was working as a clinical research fellow at the centre for CV genetics headed by Steve Humphries at the time of publishing. This article was published in the Journal of Internal Medicine, which is ranked 13/103 in the 2004 ISI Journal Citation Reports category medicine, general and internal. **2.) Beaglehole & Magnus (2002) The search for new risk factors for coronary heart disease: occupational therapy for epidemiologists?** This is a review and discussion piece concerning CHD risk factors and public health strategy. Robert Beaglehole is a well-known public health physician, currently working as an adviser to the World Health Organisation. Paul Magnus, perhaps less well known internationally, is the medical advisor to the Australian Institute of Health and Welfare. The article was published in the International Journal of Epidemiology, which is ranked 7/93 in the public, environmental and occupational health category. The analysis also looks at the ensuing responses and discussion this article prompted. This was unusual in that disagreements are made explicit in the texts. It thereby casts into sharp relief the ambiguities involved in the construction of medical knowledge. **3.) Lenfant (1999)**

**Conquering cardiovascular disease: progress and promise and 4) Beller (2001) Coronary heart disease in the first 30 years of the 21st century: challenges and opportunities.** These authors were respectively Director of the American National Heart, Lung and Blood Institute and President of the American College of Cardiology at the time of writing these pieces. Lenfant's editorial was written to mark the fiftieth anniversary of the Institute and Beller's lecture to mark the turn of a new century. Both these texts very clearly provide the authors a space to create an account of the current field and envision the future prospects. Lenfant's article was published in the Journal of the American Medical Association, ranked 2/103 in the category medicine, general and internal. Beller's article was published in Circulation, which is ranked 1/71 in the category cardiac & cardiovascular systems. Compared with the first two papers, the section of the analysis concerned with Lenfant and Beller is relatively short, reflecting the relatively shorter discussion of the causes and prevention of CHD in these papers.

#### **Publications of professional members of HEART UK**

HEART UK is now the professional organisation for a significant group of biomedical professionals involved in research concerning CHD and lipid disorders in the UK. Furthermore, it is firmly connected to the Simon Broome Register Group. This semi-autonomous research group, which is now administered through HEART UK, was formed around a national register of patients with hereditary lipid disorders to which a large number of lipid clinics contribute. Research concerning the register is undertaken by the Simon Broome Register Group Committee. It has been influential in the diagnosis

and management of FH, providing one of the internationally recognised sets of diagnostic criteria for FH (Austin et al., 2004) and epidemiological evidence demonstrating the efficacy of lipid lowering therapy in reducing CHD in people with FH. Biomedical professionals involved with HEART UK and/or with the Simon Broome Group are an important subset of professionals involved with CHD and lipid disorders. The aim of this part of the analysis was to look at what aspects of CHD and FH aetiology this body of professionals contribute to through their research activities, drawing on the framework identified in the initial analysis.

HEART UK has several hundred professional members and some 26 professionals are either trustees of the organisation or sit on one of its subcommittees. The analysis focuses on the recent publications of the individual members of HEART UK's Research Committee and of the Simon Broome Register Group Committee. They include 14 people in total, 10 clinicians and four scientists, with three of the clinicians sitting on both of the committees. The full details of the professionals and their publications is provided in Table 4.1 in Chapter 4. The rationale for choosing these particular professionals was because they are members of the two committees within HEART UK that have a clear research remit. In their positions as members of these committees they influence the research undertaken by HEART UK and by the Simon Broome Register Group. They are, therefore, likely to both have an interest in research matters and represent the different research interests of the professional members of the organisation. Furthermore, judging by their publications, résumés and performances at the HEART UK conferences, these

groups appear to include some of the most influential biomedical professionals in the field of lipidology and/or CHD risk prediction in the UK.

An author search was undertaken in September 2005 in Medline for each of these members, limited to publications from 2000 onwards. The analysis considered titles and abstracts, where provided, of all publications, including research and commentary pieces<sup>6</sup>. A total of 330 papers were included in the analysis. Medline provided abstracts for 80 per cent of these papers, and titles only for the remaining 20 per cent. The titles and abstracts of papers presented at the HEART UK Annual Medical and Scientific Meetings in 2003 and 2004 were also considered in this analysis, a further 59 papers. The analysis focuses on enumerating the main areas of interest of these researchers and how these relate to the aetiological models described in the analysis of commentary papers. This is not a detailed analysis of arguments and strategies, but a broad brush approach that builds on the framework provided by the initial analysis. It provides a quantitative element by showing the main aspects of CHD aetiology this particular group of researchers contribute to, and enumerating the number of papers they have written that focus on genetic aspects of CHD and the other aspects of CHD aetiology identified in the initial analysis.

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<sup>6</sup> Steve Humphries, a member of the Simon Broome Register Group, has a prolific output, with 185 publications listed just in the years from 2000 onwards. It was necessary to limit his papers to a manageable number. Therefore the analysis includes his papers from two years, 2000 and 2005, only, which includes 45 papers.

### **Publications of the Simon Broome Register Group**

The third set of biomedical papers analysed are the publications of the Simon Broome Register Group (Humphries et al., 2005; Huxley et al., 2003; Neil et al., 2003, 2004, 2005; Scientific Steering Committee on Behalf of the Simon Broome Register Group, 1991, 1999). As I have already argued, in the UK context, this is a very important group concerning the diagnosis and management of FH. It has provided a set of widely recognised diagnostic criteria for FH and important epidemiological data concerning mortality rates and treatment of the condition. The analysis focuses on how the aetiology and classification of FH are constructed within these publications and particularly on the place of DNA-based information in these constructions. This is compared with the constructions within a small number of commentary papers written by individual members of the group. Like the initial analysis of the four commentary papers on CHD, this analysis again involves a close reading of the arguments and evidence used in these papers to show where differences and ambiguities exist and how these are managed. It again shows how these articles are used as spaces in which to corral evidence to support particular arguments and envision specific futures for the diagnosis and management of FH.

### **3.4 ETHNOGRAPHIC WORK WITH HEART UK**

My original plan was for this part of the research to focus on the Family Heart Association (FHA). This organisation, founded in 1982 by patients, was the main patient-focused charity concerned with inherited high cholesterol. In addition, I intended to include some analysis of two further and contrasting

patients' associations concerned with CHD. During the preparatory phase of the research the FHA merged with the BHA. This necessitated a rethink concerning the questions to be addressed and the research design. As a result of the merge and in combination with reflections about the lack of detailed analysis of CHD aetiology, a new area of research concerning constructions in the biomedical literature had been added to the research design, as discussed in the previous section. Because of this additional research area and the additional questions that opened up in connection with the forming of HEART UK, it no longer seemed feasible to pursue the research on the two further patients' associations. This section of research therefore focuses entirely on HEART UK.

HEART UK remains a relatively small health charity. It currently has seven members of staff and a membership made up of both patients and professionals. Responsibility for the activities of the charity is divided between the staff, the board of trustees and a series of subcommittees with responsibilities for overseeing areas such as patient services, medical and scientific matters, and the finance and general administration of the organisation. The overall remit of the charity has altered due to the merge, now including greater emphasis on professional education, dissemination and research. Nevertheless, a central concern remains supporting and representing patients with familial hypercholesterolaemia.

The aim of this research with HEART UK was to see how the organisation constructs FH and CHD and the way these constructions are embodied in and

reinforced by its activities. Previous research suggests that different sections of patients' associations may have different priorities and ways of constructing health problems. Williams (1989), for example, showed there may be differences between discourses at the local and national level and Stockdale (1999) highlighted the existence of tensions concerning priorities, between focussing on the immediate care of members and focussing on research. These differences may or may not map onto differences between lay and professional priorities. This research was, therefore, concerned with the possibility that different people or sections within HEART UK may construct FH and CHD differently. Related to this, and in view of the merger, the analysis was also concerned with how expertise is conceived within the organisation and the kinds of relationships vis-à-vis expertise that it embodies. The research involved three main areas of activity: observation of the public events of the organisation; analysis of documentary material produced by the organisation; and interviews with members of HEART UK. The first two of these are discussed in this section and the interviews are discussed separately.

### **Access**

I made contact with the FHA at the outset of the research, as part of putting together the research proposal for my studentship application. At that stage, the organisation expressed support, in principle, for the research. The organisation has acted as both a research site and a source of information in developing the research plan. My main point of contact has been the principal nurse advisor. She provided preliminary details of the structure and organisation of HEART UK and also filled in details about how services for

people with FH are organised. I requested that the administration provide some written approval for the research involving HEART UK and this was again fielded by the principal nurse advisor. I also made contact with the Chair of the patient services committee, who agreed to facilitate contact with patients/lay people who are active in the organisation.

A couple of months after approval had been provided, I received an e-mail saying that one of the professional trustees had raised some concerns about the research when it was discussed at a trustees' meeting. In essence, he was concerned that I would judge HEART UK's activities or put them in a bad light. As Hammersley & Atkinson (1995) note, gatekeepers approach research with a set of expectations about the identity and intentions of the researcher and often adhere to a model of the researcher as either 'expert' or 'critic'. It is not unlikely for gatekeepers to associate research with evaluation, as in this case. The matter was resolved by me e-mailing the trustee. I provided details of the project and its main aims, and reassurances that the research was not controversial, and that it was not my intention, nor was I qualified, to judge HEART UK's activities. This exchange flags an important issue concerning the research on HEART UK. Because this is a small and identifiable charity, which is to some degree reliant on the maintenance of a particular image for its funding and support, I had to think carefully about how to present data on the organisation and about the nature of my relationship to the organisation. This is discussed in greater detail in the section concerning ethics.



## **Observation**

Observation is often considered the ‘gold-standard’ for qualitative research (Murphy & Dingwall, 2003). Due to the limitations of the doctoral research and the number of other aspects of the research I planned to undertake, and also due to the nature of the work of HEART UK, which includes a diverse set of activities in a variety of locations, extensive or extended ethnographic work was not feasible. Nevertheless, I planned to undertake a limited amount of observational work. It was not clear at the outset what this might include, although I planned, as a minimum, to attend public events of the organisation. Initially, I thought it might also be possible to attend some local groups and talk with local members. However, it transpired that there are currently no local support groups. The observational part of the research, therefore, included attendance of the HEART UK Annual Medical and Scientific Meetings in 2003 and 2004 and the patients’ workshops that run along side this meeting, the AGMs of 2003 and 2004 and the Annual Members’ Day in 2004 (there was no members’ day in 2003). I also visited the offices of HEART UK.

These activities were important both for the research process, in terms of becoming familiar with the activities of HEART UK, identifying key people within the organisation, negotiating access and maintaining good relations, and because of the interesting data gathered at these visits. Attendance at the earlier of the medical and scientific meetings, for example, provided an opportunity to identify some of the important people within the field and get a feel for the work of HEART UK more generally and the visit to HEART UK’s offices provided an opportunity to gather materials such as information leaflets,

back copies of the organisations bimonthly magazine, the 'Digest', details of the committee structure and contact details of the committee members, and to meet and talk with the administrative and health staff. This eased further contacts and requests for information later in the study.

At the public meetings, I made notes on the arguments and evidence enrolled in presentations and discussions, the number and type of attendees, the questions that lay members and professional members asked and how these were answered. I also reflected on the way the meetings were organised and run and the types of interactions this elicited. I endeavoured to keep a clear distinction in my records between observations of what was happening and what was said, in concrete terms, and my reflections and comments about the possible meaning of this. After each meeting or visit I compiled summary notes of my main observations and thoughts, which fed into my developing ideas for the analysis. The field notes were analysed in terms of how CHD and FH are constructed, looking particularly at the types of priorities and issues dealt with at the meetings and the way they were discussed. They were also analysed in terms of the role of lay people at the meetings and the relationship between lay and professional members demonstrated.

### **Documents**

HEART UK produces a large amount of written materials that provide a source of data about its activities, priorities, history and administration. Before providing details of the analysis undertaken, it is necessary to consider the status of such documentary material. Murphy & Dingwall (2003: 54) argue

that organisational documents sit somewhere between observation and interviews in as much as they are a source of 'given' data, in other words the data is not specifically created for the research, but involve the 'artful reconstruction' of events, as well as being part of the events of the organisation. Such documents are concerned with the self-presentation of the organisation to itself and to others, and should not be treated as providing unequivocal evidence of what they report (Atkinson & Coffey, 1997). Rather, they provide evidence of what the organisation would like to be thought of as doing (Murphy & Dingwall, 2003). The documents included in this analysis were treated as both providing evidence of the activities undertaken by HEART UK, although with caution, and as a site where particular versions of CHD and FH and particular versions of HEART UK are constructed. This discussion of how to understand documentary material anticipates, to some extent, the discussion of the status of interview data provided in the next section.

The following documents were collected: an almost complete set of AGM minutes for FHA/HEART UK from 1984 onwards, annual reports for 2003 and 2004, a complete set of patient information leaflets, copies of the 'Digest' from October 2002 onwards and other documents, including pages from the website and articles from newspapers and journals. All these materials provided useful contextualising information about the organisation and its activities. Detailed analysis was undertaken of the patients' information leaflets and of the annual reports/AGM minutes, mainly because they were two discrete and largely complete sets of data of manageable proportions.

The analysis of the patients' information leaflets focussed on the elements of CHD and FH aetiology that are present and absent in these leaflets and the way these elements are constructed. This analysis follows on from the analysis of biomedical constructions of FH and CHD, drawing on the framework of aetiological elements this identified.

The AGM minutes and annual reports were analysed in terms of the focus of the stated aims, priorities, plans and activities. The key aspect of the analysis was to look at the distribution of focus on FH, CHD and genetics, and to look for indications of the role of lay members. These documents also provided a source of information about the people who have been involved and a way of tracking changes in personnel. The analysis recognises that the AGM minutes and annual reports include an element of image management, and that they are likely to talk up the achievements of the year and set an aspirational vision for the coming year that may or may not be realised. They provide the public face of the aims and activities of the organisation. In terms of concrete activities and events, I tried as far as possible to draw on multiple sources such as press releases, the interviews, the 'Digest' and commentary in the biomedical literature. There were, however, references to a number of planned activities in the minutes that I did not come across elsewhere. These were read as indicative of the aspirations of the organisation rather than necessarily direct evidence of the activity.

### **3.5 INTERVIEWS WITH HEART UK MEMBERS AND LIPID CLINIC PATIENTS WITH FH**

#### **Why undertake qualitative interviews and what do they tell us?**

Although observation is generally seen as the method of choice in qualitative studies, there are a number of arguments in favour of interviews, and qualitative interviews in particular. The main reasons in this study were practical and pragmatic ones. Some research topics may not be amenable to observation, for example where there is no specific physical location for the topic in question, or where relevant events occur sporadically or in a private domain such as family life, where access is likely to be problematic (Bryman, 2001; Murphy & Dingwall, 2003). There may also be a pragmatic angle. For example, it may be easier to negotiate access for interviews than for more protracted observational work. My decision to undertake interviews was based on just such practical and pragmatic reasons. I have already suggested that HEART UK's activities are relatively dispersed and, that due to the constraints of doctoral research, observation over a long period of time was not feasible. 'Ordinary' FH patients are a diffuse set of people who cannot be thought of as a social group. There is little opportunity to observe how individual patients construct their condition in an organised setting. To observe the relevant actions and interactions in their everyday lives, such as in their home, at work, with family or in their social lives would be lengthy and intrusive.

Other reasons in favour of qualitative interviews include the argument that certain phenomena such as thoughts, intentions and the meaning people attach to events cannot be directly observed (Murphy et al., 1998). It is also argued

that in comparison with more standardised approaches, qualitative interviews are more flexible, so that one is able pursue interesting aspects that arise during the fieldwork period, rather than having to know in advance the important or interesting aspects of the area being researched. These arguments could also be used to justify my choice of interviews.

However, the status of interview data, i.e. the types of claims that can be made using this data, is highly contested. The main point at issue is the degree to which interviews are thought to provide access to external reality, e.g. the actions people take as a result of a diagnosis of FH, or internal reality, e.g. their ideas about disease categories and their motivations for their actions.

Silverman (1985: 15-16, in Murphy et al., 1998: 105) has argued that interviews:

‘provide idealized accounts of attitudes and behaviours which, because they are rationalisations have an uncertain relation to actual situations’.

Studies that combine observation and interviews have highlighted the potential disparity between observed interactions and people’s accounts of these interactions (see for example Allen, 1997; Stimson & Webb, 1975). The argument that interviews allow access to interviewees’ thoughts and perspectives (i.e. an internal reality) is also problematic. Murphy & Dingwall (2003) argue that there is a tendency to use such data as evidence to explain why people behave as they do, and associate this with two problematic assumptions. First, they argue that such a strategy is based on:

‘the assumption that inside informants’ heads, there are stable meanings attached to an event or experience that the interviewer must ...uncover. This approach fails to recognize the ambivalence characteristic of much of our thinking’  
(ibid: 94).

This leads them to the second problematic assumption, that the meaning and motivations given by interviewees can be seen as a direct representation of the mental states underlying their actions.

These issues concerning interpretation lead some analysts to favour an approach to analysing interview data that explicitly recognises the interview as an *example* of social interaction. It is then seen as an event that occasions ‘impression management’ guided by ‘a dance of expectation’ (Dingwall, 1997: 56). To put it simply, what we say at interviews depends on who we think we are talking to and in what situation. They should be seen as events in which shared cultural understandings of a particular phenomenon are reproduced and the focus of the analysis should be on what people are *doing* with their talk.

They are:

‘occasions on which informants are called upon to offer “accounts” for their actions, feelings, opinions and so on. In providing these accounts informants seek to present themselves as competent and, indeed, moral members of their particular communities. Interviews are occasions for informants to display themselves as adequate parents, good patients, well-informed citizens, responsible adults, and competent professionals-or to produce socially acceptable explanation of their failure’  
(Murphy & Dingwall, 2003: 95-96).

In this way, interview accounts could highlight the thoughts and actions deemed to be appropriate to the competent FH patient or HEART UK trustee. Not all authors, however, have abandoned the possibility that interview data can say anything about the interviewees’ external or internal reality. Hammersley and Atkinson (1995:126), for example, suggest that informants’ accounts can be analysed in terms of both ‘information’ and ‘perspective’, arguing that participants’ knowledge can be treated ‘as both resource and

topic'. These arguments about the status of interview data continue. This discussion has highlighted, in any case, the importance of taking into account the context of the interview in analysing such data.

In my own study the interviews with HEART UK members were seen as fulfilling two functions. First, they provided information about the activities and policies of HEART UK. As discussed in the previous section, combining data from the interviews and documents helped to provide a more comprehensive picture of the organisation's past and present activities. Second, the interviews were analysed in terms of 'perspective'. The analysis focuses on the types of constructions of FH and CHD that are embedded in the interviewees' accounts of the causes of these conditions, the activities of the organisation and its priorities. The focus here, for example, was not so much on what activities are undertaken, but which activities and topics are discussed by interviewees and which are not discussed, or the way certain activities are foregrounded.

In analysing these interviews I tried to keep in mind the way the context of the interview may have shaped the accounts provided. For example, I was aware that there may be some incentive to project a positive or successful image of the organisation and that the interviewees were used to representing their organisation and giving media interviews. It was particularly interesting to see how they approached the research interview. Only one of these interviewees seemed to provide an 'official' account. The others were more personal



accounts, providing an individual perspective in some way, although, like every encounter, they were also shaped through ‘impression management’.

In terms of the interviews with FH patients, I have tried to avoid making assumptions about the veracity or otherwise of interviewees’ reported actions or thoughts. It is not that there are any particular reasons to believe or doubt these accounts, but the emphasis was more on what they indicate about the normative frames associated with CHD, raised cholesterol and genetic disease. So for example, the section of the analysis concerned with telling relatives about the diagnosis is not so much concerned with which relatives the interviewee *really* talked to about the diagnosis, but with the way telling was or was not framed as an obligation through their interview talk.

### **Interviews with HEART UK members and staff**

#### *Sampling and recruitment*

My intention was to undertake between 10 –15 interviews with a mixture of staff, professional members and lay members of the organisation. Interviewees were mainly drawn from the staff, trustees and members of the sub-committees of the organisation. At this time, there were 7 members of staff, a total of 26 professional members who were trustees and/or committee members and a total of 5 patient members who were trustees and committee members. I had planned to include some lay members who were active at a national level and some at a local level. As already noted, it transpired that there are currently no official local support groups affiliated to the organisation. I, therefore, asked

the chair of the patients' services committee if she could suggest any further patient members who were particularly involved in the organisation.

Potential interviewees were invited to participate by letter or email and this was accompanied by an information sheet. Copies of these documents are provided in appendix 2. Everyone approached agreed to participate with the exception of one trustee who declined on the basis that he had actually just retired from the board. A total of 10 interviews were undertaken. This includes five interviews with lay/patient members and five with staff and professional members. All of the interviewees had senior positions within the organisation or had taken a lead in some part of its activities. In an effort to protect the anonymity of the interviewees, I will not provide any more specific details of their role or functions within the organisation, other than to note that the lay members interviewed included both trustees and other members.

#### *Interview format and process*

Interviews were semi-structured, guided by a set of questions drawn up in advance. These focussed on the interviewees' own involvement in HEART UK and its aims, structure and activities, its history, and hopes for the future. The exact areas and questions used in each interview depended to some extent on the role of the interviewee within HEART UK and how long they had been involved with the organisation. An interview guide containing the main areas and questions drawn on is provided in appendix 3. A few specific areas of questioning have been omitted from this, where they provide large clues as to the identity of specific interviewees.

All the interviewees agreed to the interview being recorded, and all were recorded digitally. Interviews lasted between 50 minutes and one hour and 50 minutes. The majority of the interviews were undertaken face-to-face. These took place in a number of settings, including the head offices of HEART UK, at one of the Annual Scientific and Medical Meetings, at the interviewees' places of work, and in a café. One was undertaken by telephone. This came about for a number of reasons. It was not clear at the outset how much involvement the interviewee had currently or recently had in HEART UK. Added to this, she was located in a different and distant part of the country to me, is retired and no longer travels to the HEART UK meetings. This meant that the most likely venue for a face-to-face meeting would be at her home. Such a meeting would have involved a long and potentially not particularly fruitful visit for me and an imposition for the interviewee. I, therefore, arranged to talk to the interviewee on the phone. Despite my initial uncertainty, it turned out to be a very important interview and worked very well over the phone. The call lasted for more than 90 minutes and, in this case, it was not difficult to build up a rapport quite quickly. It was not different in character or substance to the face-to-face interviews conducted.

### **Interviews at the Lipid Clinic**

#### *Selection and sampling*

Murphy et al. (1998) argue that there has been increasing concern with the issue of generalisability of qualitative research. Drawing on Schofield (1993), they suggest that in the process of selection: 'pragmatic decisions should be

integrated with a commitment to drawing samples in a systematic and principled way' showing concern for the typicality of the setting (Murphy et al., 1998: 93). Hammersley (1992: 88) also argues that:

'being unable to use probability methods does not rule out the possibility of making reasonable judgements about the representativeness of findings drawn from a particular setting in relation to some wider population'.

This section will discuss how the site for the research was selected and how sampling within this site was undertaken.

### Site selection

People with FH were recruited through a lipid clinic in a large city in the north of England. Lipid clinics are specialist outpatient clinics concerned with the care of people with lipid disorders. There are currently 135 clinics providing lipid services in the UK (Marks et al., 2003a) and it has been estimated that the majority of people diagnosed with FH are registered with a clinic (Neil et al., 2000). The decision to recruit through a lipid clinic was based on the premise that they are likely to provide access to relatively large numbers of FH patients and to have diagnosed patients precisely, i.e. there is less chance of people with other forms of high cholesterol being put forward into the study. By contrast, most general practices could be expected to have only a very small number of identified patients with FH. An alternative may have been to draw on the patient members of HEART UK. However, they are likely to be predominantly middle-class (see Rapp et al., 2001) and to have engaged with biomedical perspectives about FH. In comparison, lipid clinics offer a source of 'ordinary' patients (Lambert & Rose, 1996: 70).

The selection of the particular clinic was largely pragmatic, but has interesting theoretical implications. The pragmatic aspects were concerned with location and access. The clinic is one of a handful that were geographically convenient for me to reach, and it was suggested to me that the consultant physician who heads this clinic was both very approachable and research-orientated.

Furthermore there turned out to be practical benefits to this selection. The operational aspects of the clinic and its size meant that it was possible to reach a reasonable sample size within a workable timeframe at this one clinic, avoiding the costs of having to negotiate access to further clinics.

It is not clear how typical the clinic is of all FH clinics, and what difference the typicality of the clinic might make to patients' constructions of FH. There are a number of issues that may characterise lipid clinics, such as whether they undertake cascade screening or DNA diagnostics, the division of responsibility with primary care, and the size and prestige of the clinic. Marks et al.'s (2003a) census of lipid clinics suggests wide variations in the size and operation of these clinics, suggesting it may be difficult to define what constitutes a typical lipid clinic. The clinic at which the research was undertaken is large and run by a research-active consultant who is a recognised expert in the field. Furthermore, the clinic has undertaken cascade screening of first-degree relatives (parents, siblings, offspring) of FH patients for at least a decade. It has not yet been established the extent to which other lipid clinics in the UK are undertaking systematic cascade screening, although it is certainly patchy (personal communication, Dalya Marks, 23 June 2003). It is possible that the clinic is unusual in this respect and that such screening may encourage

patients to frame FH in a more genetic/hereditary way than other patients. On the basis of the size, location and prestige of the clinic, it is likely that it employs the most progressive practices and cutting-edge clinical techniques available. One can argue that, if geneticisation were likely to be seen anywhere, then it would be at such a clinic.

#### Within case sampling

Hammersley & Atkinson (1995: 50) suggest that sampling of people may be undertaken on 'fairly standard 'face-sheet' demographic criteria'. However,:

'these face-sheet categories are important only as they are relevant to the emerging analysis or to rival theories, or to ensuring representation in terms of some larger population, and they will usually be complemented by other categories of analytic relevance' (ibid: 50).

Clarity is called for in identifying which demographic and other criteria are important. Murphy's (2000) study of first-time mothers' feeding practices, for example, employs a quota sample based on two variables, occupational class and age, that had been identified as key variables in previous research.

Following Murphy's example, this study employs a quota sample on the basis of three variables indicated as important in the literature. Work by Hunt and colleagues (Hunt et al., 2000b, 2001; Watt et al., 2000) suggests that perceptions of a family history of CHD are patterned by gender, class and age. It is likely, therefore, that these may also be related to patients' understandings of FH. The possibility of a gendered approach to genetic responsibility has also been raised (Hallowell, 1999; Richards, 1996). A quota sample was, therefore, adopted for the categories of gender, class and age, with the intention

of ensuring that the sample was heterogeneous with regard to these categories.

The quota sample used the categories and groups shown in table 3.2:

**Table 3.2: proposed quota sample**

Gender	Age	Occupational class*	Planned Number
Male	18-45	Managerial, professional & intermediate	4
		Routine & manual	4
	46 +	Managerial, professional & intermediate	4
		Routine & manual	4
Female	18-45	Managerial, professional & intermediate	4
		Routine & manual	4
	46 +	Managerial, professional & intermediate	4
		Routine & manual	4

\*Categories are derived from the new National Statistics Socio-Economic Classification. Intermediate occupations include clerical and technical occupations and non-professional small employers and self-employed.

The sample for the study was limited to people aged 18 or over who had been diagnosed for at least six months prior to the interview. These selection criteria were applied because the interviews are concerned with people's everyday understandings and experiences of FH and these may take some time to become established. Furthermore, diagnosis is an exceptional time and may elicit a particular set of short-term concerns and issues. The research focussed on adults with FH for practical reasons. Other people for whom participation would be inappropriate, for example those who have serious illness, were also excluded.

### *Access and recruitment*

HEART UK provided details of several lipid clinics in my locality and the names and contact details of the clinicians who run them. As indicated in the section above, the principal nurse-advisor at HEART UK indicated that she knew one of the clinicians through the work of the HEART UK and that he was an approachable person and research-active. I made contact with this clinician at the HEART UK Annual Medical and Scientific Conference 2003, introducing myself and mentioning my proposed research. The clinician invited me to send him some project details and my C.V. and to make an appointment to meet him. He responded very quickly by post, agreeing to allow me access to his patients on the basis of the written information I had sent him, even before we met to discuss the research in detail. He has been helpful throughout, providing stewardship of the LREC application and guidance on completing the forms, and volunteered the assistance of his research nurse to help and advise with recruitment. Fortunately, she was also extremely helpful, despite her own demanding work commitments. In short, access to the clinic proved to be amazingly straightforward.

Application for approval by the relevant LREC was submitted in September 2003 and considered in November. It was granted conditional approval subject to a number of minor amendments and clarifications and full approval was granted on 15<sup>th</sup> January 2004. I attended the clinic to observe consultations during November 2003. This helped me gain familiarity with the organisation and administrative procedures of the clinic, which was useful in developing the recruitment procedures and forming ideas about the practicalities of the



interviews. I was also able to start to establish a relationship with the staff at the clinic. This period of observation also allowed me to start to become familiar with the language associated with lipid disorders and sensitised me to some of the issues surrounding FH.

The clinic runs on one half-day per week and currently has a register of more than 300 patients with a diagnosis of definite FH. Patients are seen every three months to year, depending on the stability of their condition and treatment. Patients were recruited from January to August 2004 and the first interview took place on 29 January 2004. The hospital was able to provide a clinic list on a two monthly basis and this was used to compile a list of patients eligible for the study, which included information about age, gender, occupation and length of time since diagnosis. A total of 117 people with definite FH aged over 18 were booked appointments between January and August 2004. This included approximately equal numbers of men and women. There were more older than younger people, with just over a quarter falling into the 18 to 45 age group. Occupational information was sometimes sketchy or missing and it was therefore not always possible to categorise occupational class with certainty or at all. Nineteen of the people could not be categorised on the basis of the available information. About a quarter of those who were categorised had routine or manual occupations.

Patients were selected randomly from the list to fulfil the quota sample based on age, gender and occupational background. Selected patients were sent a letter inviting them to participate in the study, which was accompanied by a

participant information sheet (these are included in appendix 4). They were asked to complete a return slip confirming whether or not they were willing to participate and whether they would prefer to be interviewed at the clinic or elsewhere. Fifty-five people were invited to participate and 32 people, or about 60 per cent, accepted. When broken down by age, gender and occupational group, it was noticeable that men were much more likely to agree to participate than women (18/24 men compared with 14/31 women agreed). The possible significance of this is discussed in the final section of this chapter concerning the limitations of the study.

A total of 31 interviews were undertaken in the time period indicated above. One interview was conducted at the interviewee's home and all the remaining interviews were undertaken at the hospital. Most of these were undertaken in the lipid clinic either immediately before or after the patients' scheduled appointment. Three interviews were undertaken in a separate location in the hospital when the patients came in to participate in a clinical trial. Four women brought relatives into the interview, on two occasions their husbands, once a daughter and another time a daughter-in-law.

#### *Sample characteristics*

The characteristics of the interviewees, in terms of the quota sample, are shown in Table 3.3, below:

**Table 3.3: Quota sample, planned and actual sample**

Gender	Age	Occupational Group*	Interviews Actual (planned)
Male	18-45	1	6 (4)
		2	0 (4)
	46+	1	7 (4)
		2	4 (4)
Female	18-45	1	4 (4)
		2	1 (4)
	46+	1	5 (4)
		2	4 (4)
<b>Total</b>			<b>31 (32)</b>

\*Occupational groups are based on the new National Statistics Socio-Economic Classification. Group 1 = managerial, professional and intermediate occupations (intermediate occupations includes clerical and technical occupations and non-professional small employers and self-employed). Group 2 = routine and manual occupations.

Table 3.3 shows that it was difficult to recruit to two cells i.e. younger men and women with manual or routine occupations. This can mainly be attributed to the low numbers of these groups of people attending the clinic rather than a lower response rate. There were only four men and one woman in these groups booked in for appointments during the seven-month fieldwork period. Two of the men and the woman were invited to participate in the study. Both the men declined and the woman agreed.

#### Demographic details

Interviewees ranged from 24 to 69 years old at the time of interview, although the majority (27/31) were aged 40 or over. There were slightly more men than women (17-14). Interviewees had a variety of occupational backgrounds.

Based on the new NSEC three-class categorisation, almost half (15/31) had

managerial or professional occupations, just under a quarter (7/31) worked in intermediate occupations including self-employed manual workers, and the remaining (9/31) interviewees had routine or manual occupations. The majority of the interviewees had children (25/31). All the interviewees classified their ethnicity as white, and the majority (28/31) as white British.

### Clinical details

According to the clinic's records, the interviewees were entered onto the clinic's register of FH patients between 1983 and 2002. This means that everyone had been diagnosed with FH for at least a year if not substantially longer at the point of the interview. Interviewees were between 10 and 63 years old when their diagnosis was entered onto the clinic register.

Interviewees identified the point at which their high cholesterol was established, which according to their accounts was often several years earlier than the year their FH was registered at the clinic. According to these accounts, the majority of interviewees found out that they had raised cholesterol in their twenties, thirties or forties.

According to their own accounts, approximately half of the interviewees had experienced some form of CHD, either heart attack or angina, and/or undergone some form of heart surgery such as by-pass surgery or the insertion of stents. As would be expected from the biomedical literature on FH, a larger proportion of the men than the women had experienced CHD (10/17 men compared with 5/14 women). There was also a marked difference in experience of CHD in the sample by occupational category, with the

managerial and professional group having less CHD than intermediate group who had less than the manual and routine group (4/15, 4/7 and 7/9 respectively). There does not appear to be any published data on rates of CHD in people with FH analysed by occupational class, but these differences follow the same pattern of CHD analysed by socio-economic class within the general population<sup>7</sup>.

#### *Interview format and process*

Interviews were semi-structured. An interview topic guide was drawn up, based on the research questions and the literature on genetic responsibility and lay constructions of genetic disease. It focused on explanations of FH and CHD, and interviewees' accounts of their experiences and actions in relation to FH. The final version of this is provided in appendix 5. Since the aim of these interviews was to consider in what ways patients construct FH as genetic, a major issue in designing the interview topic guide was trying to avoid pre-framing FH in a genetic way. For this reason the guide asks, for example, *who* patients talked to about their diagnosis, prompting for whether this included any family members, rather than asking primary questions about *which* family members they told and did not tell. A second issue was how to access interviewees' explanations of FH and CHD. Asking people directly to explain how they understand particular disease categories is problematic. They might feel like they are being tested against some standard textbook answer and may be reticent to answer because they feel that their knowledge is inadequate.

This issue is discussed by Lambert & Rose (1996), who approached this by

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<sup>7</sup> It would be unwise to make too much of these differences in view of the small sample size, but at first glance, this might suggest that the severity of CHD in FH varies with occupational class or that people enter the clinic at different stages of their FH career.

asking their interviewees how they would explain their condition to other people. Following Lambert and Rose, I included questions about the situations interviewees find themselves talking about their condition, what they say about it in these situations and how they might explain it to someone who had not come across it before.

The guide was amended on the basis of the first few interviews, with one question amended, one dropped and a new area added. For example, I initially intended to start by asking how the interviewee came to be diagnosed with FH or familial high cholesterol. I very quickly realised that interviewees did not necessarily identify with these terms and using them immediately framed the condition in a particular way. The question was therefore rephrased to 'how did you come to be a patient at this clinic'. This provided an opportunity to pick up on whatever phrase the interviewee used to describe their condition and then to use this throughout the interview.

All interviews began by dealing with the consent procedure and consent form, required as part of the standard LREC process. Participants were asked as a separate question whether they would consent to the recording of the interview (consent form is included in appendix 4). The interview then covered demographic details to confirm the interviewee's occupation and age, and to provide details of their domestic situation and ethnicity, before getting into the main part of the interview. The topic guide was then used in a flexible way, and interviews covered some or all of the areas depending on the direction of discussion and the relevance of the questions, and on any time constraints

indicated by the interviewee. Interviews lasted between 20 minutes and one and a half hours. All the interviews were recorded on a digital recorder and I also took notes. These were made to help me keep track of details, to indicate areas to return to for clarification or interest, or to record particular non-verbal information or interruptions. They also allowed notes to be kept of discussions before the tape was turned on or after it was turned off.

### **Analysis of interview data**

This section describes the process of analysis of the interview data in some detail. In principle this was a thematic analysis. In this kind of analysis, themes or categories are often said to emerge from the data, with little explication of the processes involved. As Hammersley & Atkinson (1995: 209) put it:

‘the development of analytical categories and models has often been treated as a mysterious process about which little can be said and no guidance given. One must simply wait on the theoretical muse’.

They suggest that there is a tendency to overemphasise the role of the creative imagination in this process, and to neglect the role of existing knowledge, arguing that the process of analysis relies on the existing ideas of the researcher and those derived from the literature, which are used as a resource to make sense of the data. There has been criticism of some researchers for failing to acknowledge the place of the researchers’ interests and prior knowledge in shaping analysis, as if categories occurred naturally in the data, waiting only to be discovered by an impartial analyst (see for example Bryant, 2003, January, and, for a review, see Silverman, 2001: pages 71-73).

Hammersley & Atkinson (1995) describe analysis as an iterative process which typically starts with careful reading of all the data and identification of ‘sensitizing concepts’ by: looking for interesting patterns and aspects that are surprising or puzzling, seeing how data relate to expectations, and looking for inconsistencies and contradictions. The data are then reanalysed to firm up categories and develop new ones, resulting in a range of categories which include both concrete and more analytical ones. This general description fits the process of analysis undertaken in this research.

A similar, although different approach was adopted in analysing the interview data with HEART UK members and lipid clinic patients. All the interviews were recorded on digital recorder and the audio files were transferred to computer. For the HEART UK interviews, I made detailed notes on each interview and transcribed in full sections that were particularly pertinent to my developing ideas. All lipid clinic interviews were transcribed in full. I transcribed most myself and a small number (5) of the later interviews were transcribed by someone else. I also made listening notes from each of the lipid clinic interviews. The making of these notes and undertaking the bulk of the transcription was an important starting point of the analysis process, giving me familiarity with the data. These activities were undertaken throughout the fieldwork period. I also kept a research diary in which I recorded any thoughts or questions about the research. All the notes and the diary were reviewed at regular intervals through discussion with my supervisors. This often provided an occasion to summarise my ideas about the analysis and the issues raised, creating a kind of ‘analytic memo’. At the end of the fieldwork period these



notes and memos were drawn together and used to develop two coding schemas.

#### *Analysis of HEART UK interviews*

There were two elements to the analysis of these interviews. First I drew up a list of the activities that interviewees mentioned or said they had been involved in. Activities mentioned in the written materials were also added to this list. Second, a thematic analysis was undertaken along the lines described in the previous section. Coding was undertaken on hard copies of the notes/transcripts. Because of the relatively small number of interviews and fairly circumscribed number of themes, the coding was managed by copying and pasting the coded sections into a Word document. This document was then printed off and reanalysed to look for patterns within each category.

Coding was shaped to an extent by the focus of the research questions. For example, it was clear that one of the themes of the thesis would concern the merge between the BHA and FHA, and all data pertaining to the merge was coded together. This was then reanalysed to look for the different types of explanations offered for the merge, which were further categorised using labels including *involvement agenda*, *credibility*, and *size/funding*. Some of the themes were informed by the analysis of constructions in the biomedical literature. For example, the place of DNA analysis was an important aspect of constructions of FH in the literature, and appeared also to be a relevant theme in interviews. Other ideas emerged from a more general reading of the data, looking for patterns or recurrent concepts, for example, ideas concerning a lack

of a cohesive illness identity. Thirteen categories were developed in total and these were organised under two main headings: aetiological issues concerning FH and CHD; and organisational aspects of HEART UK.

#### *Analysis of lipid clinic interviews*

In addition to compiling listening and analytic notes on these interviews, as already described, I appended each transcript with a top-sheet on which was recorded summary data about some of the characteristics of the interviewee that I thought might be pertinent to the analysis. This included gender, age, occupation, ethnicity, children, how and when high cholesterol was discovered, when FH was diagnosed, onset of CHD, whether CHD reported in family and whether physical signs of FH reported. These were entered onto a spreadsheet along with a note to help jog my memory about the key aspects of the interview, for example 'prompted by mum's high cholesterol and angina, focus on own ill health'. The data in the spreadsheet were used to tabulate summary data about the characteristics of the sample.

The main coding schema developed related to four core themes:

1. defining and explaining the problem
2. ideas about inherited disease.
3. health status now and in the future
4. health responsibilities

These themes map more or less onto the data chapters concerned with lipid clinic interviews. Themes 1 and 2 are discussed in chapter 6 - defining and

explaining the problem, and themes 3 and 4 are discussed in chapter 7 - living with FH. However health responsibilities permeate much of the analysis and are relevant to both of these data chapters.

An initial set of 54 codes was devised relating to these themes. Some of these codes related to a knowledge of the literature. For example, I was aware that the social and geographic proximity of relatives has been discussed in the literature concerning lay constructions of genetic risk. This sensitised me to this topic and I was able to recognise it in my own data. Other codes arose through the occurrence of a recurrent phrase in the interviews, for example, 'normal life'. Other areas were unanticipated. Prior to the interviews, I understood FH as an adult-onset condition. Therefore, the amount of talk concerned with the welfare of the interviewees' offspring was unexpected. This was related to several codes, such as 'caring for children', 'testing children' and 'passing it on'.

Coding was undertaken by marking up hard copies of the transcripts using a highlighter pen and annotating in the margins. Electronic copies of the transcripts were then coded in NVIVO. Further analysis of the initial codes was undertaken by printing out the data categorised in each code, reading through this data, differentiating it into a number of ideas, and marking this up on the hard copies. The use of computer-aided qualitative data analysis software (CAQDAS) has been subject to much discussion and it has been proposed that it has the potential to transform the analytic process (see Murphy & Dingwall, 2003: pages 125-127 for a brief synopsis of the arguments). My

own use of CAQDAS was intended solely as a convenient way to manage coding, data storage and retrieval. All of the procedures carried out within NVIVO could equally have been achieved using manual methods of data management (cutting up hard copies of transcripts with a pair of scissors and storing in cardboard files) and using a word processing package for the occasional word search. The choice of NVIVO was because it was available and because I was already familiar with the package from previous research.

The full coding schema was initially tested on three transcripts to check that it could be applied and see whether anything was missing. It was then applied to the full set of transcripts. There were several areas that I attempted to tabulate in order to get some overall sense of the data or distribution of ideas, with different levels of success. For example, I tried to categorise the main talk about FH into three categories – medication, diet and heredity. This information was recorded on the transcript top-sheet. It turned out that most people focussed on two or more of these at different points in the interview. This provides an indicator of the multiple ways of constructing FH, which, in fact, became a major theme of the analysis discussed in Chapter 6. Nevertheless, this categorisation was useful in identifying the small number of interviews where there was no or little talk concerning one of the categories.

### **3.7 REFLECTIONS ON THE RESEARCH DESIGN AND PROCESS**

#### **Ethical issues**

There are two aspects of this research that warrant some consideration of the ethical issues raised. These are the interviews with FH patients and the work with HEART UK, including the interviews with its members and staff.

The interviews with FH patients required approval from the Local Research Ethics Committee, which, to an extent, provided a template for the management of the interviews. Nevertheless, these interviews did not raise any particularly novel ethical issues. The recruitment and data management processes were designed to address the issues of informed consent, confidentiality and anonymity of participants and their rights of care. This meant, in practice, that potential participants were sent a participant information sheet in advance of the study, and were asked to sign a consent form, transcripts were identified using a numeric ID number, hard copies of the transcripts and identifying details were stored securely, and electronic versions of transcripts and audio-files were pass-word protected. Although conforming to the institutional demands regarding LREC approval, I recognise that these measures do not automatically ensure the ethical treatment of participants. The idea of ‘informed consent’, for example, is a thorny issue. Information provided to potential participants must be accessible and comprehensible to this audience. It would be difficult to provide a full explanation of the aims of sociological research of this nature ‘without sending informants and

cohabitants to graduate school' (Brewster Smith, 1979: 14, cited in Murphy & Dingwall, 2001: 342).

Ethical issues concerning the research with HEART UK are less clearly defined. HEART UK is a small organisation with a unique remit within the UK. The organisation would be instantly recognisable whether or not it was named in the research. It also has a small number of staff. If I had identified interviewees as staff members, readers would have been able to make a fairly educated guess as to their identity. I, therefore, decided to group staff with professional members in reporting the data. Lay members are denoted as LM1-5 and staff and professional members as S/PM1-5<sup>8</sup>. It is highly likely, nevertheless, that the interviewees will be recognisable to people within the organisation. It is notable that the professional members interviewed were drawn from the same population as the sample whose published literature is analysed. Where data pertained to a written source that is already in the public domain, I have named the authors.

Earlier in this chapter, I indicated that there was a small amount of concern within HEART UK about the research and how it will be used. I suggested that these concerns were allayed through the provision of information and reassurances about my intentions. My own view is that this thesis does not evaluate the organisation, nor is it critical of its activities. The work of the organisation is analysed to consider the concepts of geneticisation, lay expertise, biosociality and so on. However, the information I provided to the

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<sup>8</sup> The same procedures for ensuring secure data storage and protection were followed as those described for the FH patient interviews .

organisation, as part of negotiating access, was not framed in these terms. This would have been equally incomprehensible and irrelevant to the HEART UK as to the FH patients (see discussion above on the difficulties of informed consent). The research was explained as being concerned with the work of HEART UK in supporting patients and shaping developments, and with the way patients and professionals work together in the newly merged organisation (see appendix 2 for the letter of invitation and information sheet for HEART UK participants). This thesis represents the entry of this part of the research into the public domain and I cannot anticipate how it will be understood by the organisation or other audiences. The point of publication may be the time of greatest risk of harm to participants in social research (Murphy & Dingwall, 2001). I will have relatively little control over how this research is read by others. Nevertheless, I believe I have a responsibility to HEART UK to think carefully about how I write about its activities in a way that decreases the chance of this research being seen or used as an evaluation.

#### **Limitations of this study and reflections on the research design.**

*Biomedical constructions of CHD/FH* – As I have argued earlier in this chapter, there are many ways of studying biomedical constructions of diseases and the method chosen was the most accessible under the circumstances. The decision to start by analysing four commentary papers followed Hedgecoe's analysis of the rhetorical strategies used in constructing diabetes and schizophrenia (Hedgecoe, 2001a, 2002). This method has recently been criticised, in part, for the small number of articles analysed (Kerr, 2004) and my analysis could, perhaps, be open to the same criticism. Nevertheless, my

initial analysis is supplemented with a summary of a large number of papers published in a particular field, which builds on the framework developed in the early analysis.

The study provides a systematic analysis of one small area of CHD constructions, relating to the work of HEART UK and researchers connected to this organisation. The analysis is indicative of the variety and kinds of arguments that are currently enrolled in constructions of CHD, and the recent focus of HEART UK researchers, but is not an exhaustive account. The analysis does not provide a basis on which to discuss whether there is a dominant model of CHD, what this is, or how it developed. The analysis of constructions of FH focused particularly on the publications and members of one group, the Simon Broome Group, which has been important in the UK. Again, this was just one slice of the biomedical literature about FH, not a definitive account. As I have argued at the beginning of this chapter, there is almost unlimited scope for detailed qualitative and quantitative work in this area.

*Work with HEART UK* – One of the characteristics of qualitative research is that the research design is flexible and develops throughout the research process. The merge between the FHA and the BHA to form HEART UK contributed to the shift in focus of the study onto professional constructions of FH and CHD. Had the research started three years earlier, this would possibly have led to a very different study and if the fieldwork had bridged the period of the merge this could have provided a unique opportunity to observe, first hand,



the process of the merge and the events surrounding this. HEART UK is clearly in a state of flux, and rapid changes have occurred and continue to occur in its structure and activities. It must be recognised that organisations such as HEART UK are dynamic entities and that this research provides an account of one particular moment in its development.

The work with HEART UK was particularly shaped by the exigencies of the research process, being circumscribed by the timetable associated with doctoral research and the nature of the work undertaken by HEART UK. A longer ethnographic study might have provided more insights into the roles of different actors in the organisation and balance of activities between FH and CHD. It might have been possible, for example, with more time and good relations, to have negotiated access to committee and trustees' meetings. This still leaves issues concerning the large number of activities that take place 'off site' and in collaboration with others and raises questions about whether the various actors would have tolerated the researcher's presence and issues of the need to repeatedly negotiate access.

*Patient constructions of FH* - Patients with FH were recruited through one lipid clinic only, due to the practicalities of gaining access and the usual constraints on doctoral research. Although the sampling method employed was designed to obtain a diverse sample of patients attending the clinic, like many qualitative studies, questions remain about the generalisability of the findings (Murphy & Dingwall, 2003). It is possible that there are aspects of the clinic's characteristics or practice that have a particular influence on patients'

constructions of CHD and FH. Aspects that may be relevant, in the case of lipid clinics, include the size and location of a clinic, and the degree to which DNA testing and systematic cascade screening are already undertaken. However, as this chapter suggested earlier, on the basis of the characteristics of the clinic involved in the research, i.e. a large and prestigious urban clinic that operates cascade screening, one can argue that it would be one of the most likely sites where geneticisation would be evident, if it were to be seen anywhere.

One must also consider the nature of the sample that is provided by a clinic population. This is a sample of clinic attendees and three groups of people with FH are not represented here: First, there are people whose FH is managed through primary care, although Neil et al. (2000) suggest that this constitutes a relatively small proportion of those who have been diagnosed with FH. Second, there are people who may be aware of a family history of CHD or FH or have been diagnosed with FH, but who have chosen not to be in contact with health professionals regarding this or have declined an invitation to be tested. The size of this population is uncertain and accessing such a population is fraught with both ethical and practical difficulties, such as whether it is ethical to contact people who have declined service, and how to establish that there is FH in the family or that the person has FH. Third, there is a group of people who declined the invitation to participate in my research. It is possible that those who participated have a more morally coherent story or perhaps were less upset by their FH. It was noted earlier that a larger proportion of men than women accepted an invitation to participate. These observations would

certainly fit with the suggestion in the literature that genetic and wider health responsibilities are gendered. Nevertheless, there were some stories of regret or self-blame present in the accounts of the participants, in other words they are not excluded from the sample, and examples are discussed in chapters 6 and 7. Furthermore, the data do not support any obvious gendering of responsibilities vis-à-vis FH.

### **3.8 SUMMARY**

This chapter has laid out in some detail the rationale for the research design and provided an account of the methods of the study, paying attention to the sampling decisions, the conduct of the research, the nature of the data produced and the process of analysis. It has also provided a discussion of the limits of the study, which is intended to indicate the sorts of claims that can legitimately be made on the basis of the research. As Murphy et al. (1998) argue, the clear exposition of the methods of data collection is one of the main criteria by which the validity of qualitative research can be evaluated.

The chapter has shown how the original focus on patient constructions of FH was modified due to two key developments. First, this was based on my growing awareness that there was, in fact, a paucity of detailed analysis of recent biomedical models of CHD. Second, it was connected to the merge of the FHA with BHA to form HEART UK at the earliest stages of the study. These two factors contributed to the broadening of the research to include questions about the construction of biomedical models of CHD and about the relationship between lay and professional members in HEART UK. The

research involved three main areas of research: an analysis of biomedical literature, ethnographic work with HEART UK and interviews with HEART UK members and patients with FH. The following chapter presents the analysis of the first of these areas, the construction of CHD and FH in the biomedical literature.

## **CHAPTER 4: BIOMEDICAL CONSTRUCTIONS OF CHD AND FH**

### **4.1 INTRODUCTION**

This chapter focuses on biomedical or ‘expert’ constructions of the aetiology of CHD and FH, and particularly on the models supported by biomedical professionals who are involved with HEART UK. The first half of the chapter looks at recent constructions of CHD within biomedical literature, to provide an overview of this field. The aim of this analysis is to indicate the range of ideas currently circulating about the causes of CHD and the place of genetic ideas in this field. This section is based on an analysis of four published commentary papers by eminent scientists.

The chapter then turns to research connected to HEART UK. The second section of the chapter discusses the contribution of HEART UK members to models of CHD. This focuses on the research profiles of individual professional committee members, based on an analysis of their recent publications. The third section focuses on constructions of FH and is based predominantly on an analysis of the publications of the Simon Broome Register Group. This is a semi-autonomous research group, formed around a national register of patients with hereditary lipid disorders, which has been influential in the diagnosis and management of FH in the UK. The group is now administered through HEART UK. These two areas of publications represent the contribution of a significant group of biomedical professionals, who are involved in some way with lipidology, to constructions of CHD and FH.

The chapter will argue that a geneticised model of CHD exists, but that there are a number of alternative and competing models of CHD, and that genetic models were not dominant in the literatures included in the analysis. It will suggest that even in the area of FH there is some ambivalence about the utility of genetic testing and, in the case of the UK, diagnostic criteria remain largely clinical rather than genetic. This analysis leads to a discussion of the influence of different disciplinary perspectives on constructions of CHD. The chapter concludes that the data presented challenge the geneticisation thesis in a number of areas.

#### **4.2 A BRIEF OVERVIEW OF THE MOLECULAR BIOLOGY OF CHD**

Before setting out the analysis of biomedical publications on CHD and FH, this section will provide my own very brief account of current thinking on the molecular and cellular processes that lead to CHD. This is included to provide the context for and aid understanding of the following analysis.

Molecular models of CHD are concerned with the development of atherosclerosis in the coronary arteries. This means the hardening and narrowing of the blood vessels that supply oxygen and nutrients to the heart muscle. The focus of the molecular model of atherosclerosis is the development and stability of fatty plaques, or atherosclerotic lesions, which form in the lining of the coronary arteries. This is currently thought to involve three main processes:

1. **lipoprotein metabolism:** Part of the process of fatty plaque formation involves the accumulation of cholesterol within the arterial wall.  
  
Lipoproteins are the molecules in which cholesterol and other lipids are carried around the body via the blood stream. Two molecules, low density lipoprotein (LDL) and high density lipoprotein (HDL) are thought to be particularly important. LDL carries the cholesterol to the cells and tissues and HDL carries cholesterol from the tissues to the liver, where it is processed for excretion. High levels of LDL and low levels of HDL are associated with the formation of atherosclerosis.
2. **the inflammatory process:** this is also involved in the formation of the fatty plaques. Macrophages, cells involved with inflammation, are responsible for the uptake of cholesterol into the arterial wall. The inflammatory process is also involved in the stability of the fatty plaque and influences whether a plaque will rupture. Plaque rupture can cause the arteries to become blocked and result in a heart attack
3. **Coagulation or clot formation:** this can occur as a result of plaque rupture and can also lead to the arteries becoming totally blocked and to a heart attack

#### **4.3 MODELS OF CHD IN THE BIOMEDICAL LITERATURE**

This section provides an overview of current constructions of CHD by considering in detail four recent commentary papers about CHD from the fields of genetics, epidemiology and cardiology. The rationale for selecting these papers and details of the authors were discussed in detail in the previous

chapter. The papers all provide very general and broad accounts of their subject areas. They are:

1. Stephens, J.W. and Humphries, S.E. (2003), 'The molecular genetics of cardiovascular disease: clinical implications', *Journal of Internal Medicine*, 253, 120-7.
2. Beaglehole, R. and Magnus, P. (2002), 'The search for new risk factors for coronary heart disease: occupational therapy for epidemiologists?' *International Journal of Epidemiology*, 31, 1117-22.
3. Lenfant, C. (1999), 'Conquering cardiovascular disease: progress and promise', *The Journal of the American Medical Association*, 282, 21, 2068-70.
4. Beller, G.A. (2001), 'Coronary heart disease in the first 30 years of the 21st century: challenges and opportunities. The 33rd Annual James B. Herrick Lecture of the Council on Clinical Cardiology of the American Heart Association', *Circulation*, 103, 20, 2428-35.

### **A genetic model of CHD**

Stephens & Humphries (2003) is a review paper concerning the effects of functional polymorphisms<sup>9</sup> on CHD. The paper focuses on three examples of

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<sup>9</sup> *Genetic polymorphism* means gene variations. Some variations have no effect on the gene's function and may be referred to as neutral variants. Others lead to variations in the gene product and may be referred to as 'functional gene polymorphisms'. These may or may not be thought to be disease causing or pathogenic (human blood groups are probably the best known example of functional gene polymorphisms). *Mutation* also means variation in a gene. It can mean merely a random change in the DNA, but in medical papers it usually denotes a pathogenic change, and often means an inherited pathogenic change. There is no precise definition of polymorphism or precise distinction between the terms polymorphism and mutation. Use of polymorphism is often associated with more prevalent changes. It tends to be used only where a variant occurs with a frequency of at least 1 per cent. More importantly, in biomedical papers, polymorphism tends to be used for benign variations and mutation for pathogenic changes.



these, two involved in lipid metabolism and one involved in inflammation. It demonstrates a familiar narrative about a genetic future for understanding and managing cardiovascular disease. CHD is presented as a polygenic, multifactorial condition in which genetic risk results from the cumulative effect of variations in several different genes. The overall message is that understanding the role of genetic polymorphisms is key to understanding the molecular and cellular pathology of CHD, which in turn may be important for the development of new therapies, diagnostic tests and prevention strategies:

Unravelling the role that functional gene polymorphisms play in determining risk...is crucial to our understanding of the key metabolic pathways and physiology not only in the diseased, but also in the disease free state. The identification of new molecules...may subsequently lead to potential targets for therapeutic intervention. Improving our molecular understanding may also lead to the development of improved diagnostics' (ibid: 126).

At the same time the authors acknowledge the importance of the more established risk factors and a large part of the paper is devoted to gene-environment interactions, focussing particularly on smoking. Interaction is used to describe the situation where the cumulative risk of two or more factors is greater than the individual risks of each factor added together.

The introductory part of the paper sets out the importance of genetic factors in the aetiology of CHD. The key argument is that:

'functional gene polymorphisms account for much of the biological diversity in homeostatic systems. In their absence all humans would respond in an identical manner to an environmental challenge, and the risk of developing disease would be directly proportional to the environmental stimulus. We know that this is not the case. For example, some individuals exposed to cigarette smoke with an *otherwise identical risk factor profile* will go on to develop CHD, whilst others will not. Therefore, the well accepted view is that CHD is

a multifactorial disorder, with both environment and genetic factors contributing’  
(ibid: 120-121, emphasis added).

This argument hinges on the idea that it is possible to identify people with ‘otherwise identical risk factor profiles’, which implies that all the risk factors for CHD are already clearly established and that they can be controlled for. This argument could perhaps be re-written as – *there are variations in the occurrence of CHD that have not been explained yet and we attribute this remaining variation to genetic factors.* This model of CHD allows a major role for genetic variations. The authors claim this is *the well accepted* view, which closes off any scope for discussion. It is, however, not a view that is universally accepted by biomedical scientists, as the analysis later in this section demonstrates.

It is particularly striking how the paper constructs genetic differences, environmental risk factors, and the relationship between environment and genes. The authors appear to use the terms mutation and polymorphism interchangeably with the same variations referred to as both ‘minor mutations’ and as ‘functional gene polymorphisms’ (ibid: 120-121). Variations in apolipoprotein-E (APOE) gene are discussed, a gene that codes for a protein involved in lipid transport. There are three APOE variants, E2, E3 and E4, and their frequencies are given as 8, 77 and 15 per cent respectively in white populations. Through the discussion E3, the most common variant, emerges as the normal gene. The other variants are referred to as ‘single common mutations with modest impact’ (ibid: 121). Since more than 40 per cent of the population have at least one copy of the E2 or E4 variants, it could be argued

that this effectively constructs a large proportion of the population as abnormal in some way.

An interesting conception of environmental risks also emerges in the paper. Environmental challenges are said to include: ‘diet, male sex, diabetes, obesity and cigarette smoking’ (ibid: 121). It is not clear what the authors have in mind in their reference to diabetes. They may have meant it as a proxy for ‘lifestyle’ factors such as inappropriate diet and lack of exercise associated with obesity and Type 2 Diabetes. However, it could also be read as a physiological or bodily state concerning glucose metabolism. Reference to male sex, i.e. being male, certainly suggests that the environment envisaged by the authors is the biological environment of the genes or of the organs rather than the physical or social environment of the person. The authors also link environmental risks and lifestyle choices, seeming to conflate the two concepts:

‘individuals adopt a different position on the environmental spectrum of risk by the lifestyle choices they make (e.g. smoking). However, although environmental risk factors are modifiable the genetic factors are not’  
(ibid: 124).

The excerpt shows that, in this paper, one of the defining attributes of the environment is that it can be changed. It is ironic, then, that male sex is included as an environmental factor. This obvious contradiction serves to highlight the rather limited conceptualisation of environment employed.

It is not clear in the model outlined in this paper whether environment is envisaged as being able to play a role on its own, in the absence of genetic

predisposition. CHD is referred to as a 'polygenic' disease. The only exceptions discussed are cases where the disease is monogenic, involving a single rather multiple genes. The implication is that genes are always involved in CHD in some way. One of the opening statements of the paper is that:

'CHD is a complex condition resulting from numerous gene-gene and gene environment interactions'  
(ibid: 120).

This suggests that genes are a necessary part of CHD aetiology. On the other hand, the authors go on to comment that CHD may occur:

'as a result of failure at the genetic level (e.g. gene transcription) or due to an environmental exposure (e.g. smoking) or due to an imbalance between the two'  
(ibid: 120).

This implies that environment alone may cause CHD in some cases.

The paper's discussion of gene-environment interactions suggests that certain people may be exposed to a given environment that 'amplifies the risk associated with that gene' (ibid: 121). In terms of the priority attributed to these factors, this seems to privilege genes. One wonders why the environment is seen as amplifying the effect of genes rather than the gene amplifying the environment, particularly as genetic variations on their own tend to account for only very minor differences in rates of CHD. This is a matter of subtle difference and, in isolation, one might not be inclined to read too much into this way of ordering things. Nevertheless, taken as a whole, there appears to be a subtle privileging of genetic factors in this paper. There are parallels here with Hedgecoe's (2001a) analysis of biomedical discourses about schizophrenia. Hedgecoe suggests that these discourses under-specify and downplay the environment and prioritise genetic factors to the extent that a

‘genetic baseline’ is seen as necessary for the causation of the disease. The conclusions here regarding the notion of a ‘genetic baseline’ for CHD are more tentative. The analysis suggests that genetic variations may be seen as having a necessary part in CHD causation.

Overall, Stephens and Humphries’ account of the causes of CHD does seem to attribute a large role to genetic differences between people, creating new categories of biological difference. The interchangeable use of the terms variations, polymorphisms and mutations tends to pathologise some of these new categories of people. It also presents a rather incomplete view of environmental factors in which the social and physical context are absent. Further, their discussion suggests that new genetic knowledge is imperative for progress in understanding CHD and will be useful for treating the disease. In other words, in terms of Lippman’s definition, this is a thoroughly geneticised view of CHD. It is, however, not the only available model of CHD, as the following sections demonstrate.

### **Epidemiology fights back**

This section considers the article by Beaglehole and Magnus (2002a), provocatively entitled ‘The search for new risk factors for coronary heart disease: occupational therapy for epidemiologists’? The article was accompanied in the same issue by four commentaries (Greenland et al., 2002; Law, 2002; Marmot, 2002; Nieto, 2002) and a response by the authors (Beaglehole & Magnus, 2002b). This dialogue provides an interesting window on current debates about CHD within epidemiology. It provides an entirely

different perspective on risk factors for CHD and demonstrates that genetics is just one of a number of emerging and contested areas of interest in CHD.

Beaglehole and Magnus's (2002a) basic argument is that the major risk factors for CHD have already been established, i.e. high blood cholesterol, high blood pressure, cigarette smoking and physical inactivity, and that these can be attributed to economic, social and cultural factors:

‘The underlying causes of the CHD epidemic are the society-wide economic and cultural factors that determine whether a diet with a high proportion of saturated fat and low in antioxidants becomes widespread... The prevailing social and economic conditions are also responsible for the emergence and widespread distribution of other important contributing causes: tobacco smoking, physical inactivity and other inappropriate aspects of diet, with the latter two interacting to produce excess weight and high levels of blood pressure. These major causes have a close and precisely defined (proximal) relationship to the CHD epidemics and are well established scientifically’  
(ibid: 1118).

It is notable that what is *well established scientifically*, in other words, what every one knows about CHD in Beaglehole and Magnus's world already differs from the *well accepted view*, in Stephens and Humphries' world.

Beaglehole and Magnus argue that the search for new risk factors is partly based on researchers underplaying the role of the main factors outlined, as a way of justifying new research. They argue that there is a widely accepted ‘myth’ that the established CHD risk factors explain only half or less of the occurrence of CHD, whereas in their view these major risk factors explain at least 75 per cent of new cases (ibid: 1119).

They go on to discuss the new risk factors, which they divide into six main groups:

- Thrombotic factors and the effect of biochemical markers
- The role of inflammation and infectious agents
- The influence of early life exposure
- The contribution of multiple genes
- Oestrogen deficiency
- The role of the psychosocial environment

(ibid: 1119).

The authors evaluate these factors in terms of their potential contribution to prevention of CHD and the degree to which they contribute to explaining 'CHD epidemics'. They are particularly critical of genetic research and better disposed towards some of the other factors. In the end, however, all the factors are variously dismissed due to lack of conclusive evidence or because their potential contribution to improving population health is doubted. The majority of funding, they argue, should be directed to prevention strategies at the primary and population levels, including structural issues such as the salt content of processed food, the environmental factors leading to lack of physical activity and tobacco policies. In an argument echoing Lippman's, they conclude that preoccupation with identifying high risk groups and individuals reflects the dominant health paradigm, which, they argue:

'supports an individualistic approach to health improvement and ignores the wider social and economic determinants of the health of populations'  
(ibid: 1121).

It is notable that, in contrast with Stephens and Humphries, environmental factors are framed in terms of the cultural, economic and social factors that lead to CHD risk, rather than as *life-style choices*.

Beaglehole and Magnus's argument is essentially that there is a gap between current knowledge and practice regarding population levels of the established risk factors and there should be more focus on prevention strategies at this level. None of the four commentaries that follow the article take issue with these main arguments, but contest Beaglehole and Magnus's arguments concerning the new or emerging risk factors and their potential contribution. The commentaries make three main types of arguments: they propose alternative aims for epidemiological research, dispute the evidence proposed by Beaglehole and Magnus and provide counter evidence and arguments to support the 'new' risk factors.

One of the main arguments they make concerning the aims of epidemiological research, in essence, reflects Rose's (1985) discussion of the two ways of understanding the aetiology of disease. These are seeking the causes of cases, which focuses on the individual, and seeking the causes of incidence, which focuses on the population. While Beaglehole and Magnus are solely interested in population levels of CHD, the commentators propose that there are other legitimate aims for epidemiological researchers. One of these is to explain why certain groups or individuals are more or less likely to get CHD. Marmot (2002: 1124), for example, is particularly interested in why rates for different groups of people vary so greatly, asking why:



‘among people more or less equally exposed, there remain such marked differences in the rate of occurrence of CHD’.

His own work is concerned with explaining the social gradient in CHD and has particularly focused on psychosocial factors. It is notable that Stephens and Humphries employed the idea that only certain individuals with a given level of the known risk factors go on to get CHD as an unequivocal argument for genetic influences, implying that there was no other possible explanation. Yet, the same argument can and is employed to support research into other risk factors.

Commentators are critical of the idea that 75 per cent of CHD is explained by the established risk factors. They also question the evidence provided by Beaglehole and Magnus in their evaluation of the emerging risk factors, criticising both individual papers cited and the types of evidence drawn on in general. Counter evidence and arguments are made in support of psychosocial factors (Marmot, 2002), infections or inflammatory processes (Nieto, 2002; Greenland et al., 2002), early life exposures and the lifecourse approach (Law, 2002; Greenland et al., 2002) and thrombotic processes (Greenland et al., 2002). What these discussions make clear are that there are a range of proposed risk factors for CHD and they bring to the surface the uncertainty and contingency of scientific claims. What is notable is that no defence of multiple gene influences is made. In these discussions, in any case, genetic explanations appear to be low on the agenda.

These discussions also illustrate a point made in the literature review, that Lippman’s concerns form part of a wider critique concerning individualistic

approaches to health and a lack of attention to the wider determinants of health and illness, which can be found in biomedical discourses as well as in social science discourses (see Aronowitz, 1998). It is interesting that while Lippman's criticism is directed towards geneticists and their allies, both Beaglehole and Magnus, and Marmot make antagonistic comments about the role of cardiologists in individualising prevention strategies for CHD:

‘without strong epidemiological input and leadership, cardiologists will continue to dominate the prevention debate and individual approaches will remain the priority’  
(Beaglehole & Magnus, 2002b: 1134).

The next section suggests that clinicians may tend to focus more on individual-level prevention. This does not mean a wholesale adoption of genetic discourses.

### **The clinicians' view**

This section considers papers by two clinicians, Lenfant and Beller. Lenfant's (1999) editorial piece sets out to provide an overview of recent progress and issues in cardiovascular disease research. He makes three main arguments in relation to coronary heart disease: (1) there is still work to be done concerning the established risk factors, for example, there is a lack of knowledge about how obesity acts as a risk factor and how it leads to diabetes, (2) molecular biology and molecular genetics may offer major improvements in the field of cardiovascular disease, and (3) there is a need for better application of the established interventions, in other words that there is a gap between knowledge and practice.

Lenfant (1999: 2069) is optimistic about the role of genetics:

‘On the eve of the next millennium it is safe to predict that the importance and application of molecular genetics and functional genomics will play major roles in the further improvement of cardiovascular health’.

Although he makes reference to genetic polymorphisms, equally he includes discussion of developments in molecular biology concerned with the role of various molecules and cellular processes in the development and stability of plaques and the function of the arterial lining. It is important to note that such developments may be concerned with understanding the molecular processes that lead to CHD, in general, and with finding new ways of assessing the risk of and progress of disease, based on this knowledge. Such research is not necessarily related to identifying genetic differences between people.

A major part of Lenfant’s argument is that there is a problem translating research results into lifestyle changes, public health interventions and clinical practice, i.e. a gap between knowledge and practice, and highlights this by discussing evidence concerning poor use of drugs for blood pressure and poor ‘compliance’ with blood-pressure treatments. He concludes that although molecular biology and genetics offer exciting and useful prospects, doctors should not forget about the ‘more mundane’, but effective methods like lowering blood pressure, decreasing obesity and physical inactivity and the appropriate use of established therapies such as  $\beta$ -blockers and aspirin:

‘The real challenge for the new millennium may indeed be to strike an appropriate balance between the pursuit of exciting new knowledge and the full application of strategies that already are known to be extremely effective, but considerably underused’  
(ibid: 2070).

Beller’s (2001) review, entitled ‘Coronary Heart Disease in the First 30 Years of the 21<sup>st</sup> Century: Challenges and Opportunities’, would seem to provide a

perfect opportunity to focus on predicted developments in, and applications of, cutting edge technologies such as genetics. However, the major challenges for CHD prevention and treatment are framed in more prosaic terms. Discussion focuses on three main factors (1) an increasing proportion of older people in the population, (2) an 'epidemic of type 2 diabetes' and linked to this (3) an 'obesity epidemic'. Beller's main response to these challenges is to draw on a discourse of a gap between knowledge and practice. He suggests that older people are already under-treated, arguing that they are not benefiting enough from '*proven* diagnostic and therapeutic strategies' (ibid: 2429, emphasis added). The paper relates diabetes and obesity in part to genetic factors. Nevertheless, the main thrust of the discussion about these conditions is concerned with physical inactivity and poor diet. Beller calls for greater emphasis on prevention in these areas, stressing the responsibility of cardiologists, parents and educators. In contrast to Beaglehole and Magnus, with the exception of commenting on education policy concerning physical education programmes, these are largely taken as individual problems requiring individual actions.

For the future, an array of technological developments, both biological and mechanical, in all areas of prevention and treatment are predicted. Beller mentions the prospect of genetic screening to identify people at risk of diabetes and CHD, but he appears to link this solely to prevention for people at high risk of early CHD. In other words, this is a fairly limited role for genetic developments, rather than the more global role suggested by Stephens &

Humphries (2003). In conclusion, he concurs with Lenfant and urges us not to forget the:

‘less costly, low-tech interventions [that] have already proven effective in preventing CHD and its complications’  
(Beller, 2001: 2434).

In sum, Lenfant and Beller display some enthusiasm for developments in genetics. Again, however, this could certainly not be thought of as **the** dominant theme in their accounts of the future. Their message is that health improvements may result from developments in genetics, but more importantly, could be accrued through better implementation of a raft of established low-tech, or even no-tech, interventions i.e. through better application of what is already known.

### **Summary and Discussion**

The analysis of Stephens and Humphries (2003) demonstrates that there certainly is a genetic model of CHD that has many of the elements of the geneticised discourse proposed by Lippman. The account of CHD provided by Stephens and Humphries privileges genetic causes while detracting from other causes and creates new categories of biological normality and abnormality. It also suggests that new genetic knowledge is imperative for increased understanding and improved treatment of CHD. However, the analysis of the four commentary papers taken together suggests that genetics is just one of a number of streams of discourses about CHD. One stream continues to focus on the established risk factors and includes emphasis on population trends such as levels of obesity and diabetes, or an ageing population. This was connected to a recurrent argument that there is scope for major improvements, whether at

the population or individual level, if only better use was made of current knowledge and interventions. This is far from the argument that health improvements will and can only be achieved through genetic research anticipated by the geneticisation thesis.

Attempts to explain variations which cannot be accounted for by the established risk factors (although how much is already explained is contested) involve a range of risk factors of which genetics is just one. Some of these are concerned with the molecular biological processes involved in atherosclerosis. There is interest in establishing whether certain molecules involved in inflammation and thrombosis could be used as new markers of disease progression and whether they play a role in causation. It must be noted that this interest in new disease markers is not necessarily related to a genetic account based on inherited differences between people. In other words, interest in molecular biological processes should not be conflated with interest in genetic variations between people.

Overall, the analysis suggests that there is significant heterogeneity in biomedical discourses about CHD and that there are currently a number of ways of understanding the role of genetics. It can be seen as a unifying genetic model, an explanation for a subset of early ('premature') CHD, or a general recognition that CHD can run in families. Genetics certainly did not represent **the** dominant discourse. The analysis also demonstrates that there are a number of disciplines with an interest in CHD and has hinted at boundary work (Gieryn, 1983), at least by epidemiologists. This is evident in their claims for

control over prevention strategies for CHD and admonition of cardiologists for promoting individual-level approaches to prevention. This suggests that Lippman's claims about the power of geneticists to condition how a host of diseases are viewed is, at the least, overstated. Furthermore, the analysis suggests that disciplines may differ in their orientation towards individual or population/strategic level interventions, reflecting different ways of constructing responsibility for health and illness. Nevertheless, those with a more individualistic orientation do not necessarily prioritise genetic discourses about CHD.

#### **4.4 HEART UK'S CONTRIBUTION TO MODELS OF CHD**

This section discusses the research undertaken by biomedical professionals involved with HEART UK. These are an important subset of professionals involved with CHD in the UK. One can argue that their research activities both reflect the priorities of the organisation, or at least those of its professional members, and potentially contribute to particular aetiological models of CHD. It will suggest that researchers' interest in genetics is mainly concerned with specific hereditary lipid disorders rather than in polygenic influences as a more general risk factor for CHD, and that their main focus was concerned with elucidating the molecular pathways associated with atherosclerosis.

The analysis in this section focuses on recent publications, since 2000, of the individual members of HEART UK's Research Committee and of the Simon Broome Register group. The rationale for selecting these two groups was outlined in the previous chapter. To recap, these are the two committees with a

clear research remit within HEART UK. It, therefore, seemed likely that the committee members would have an interest in research matters, and would represent the different research interests within the organisation. The analysis involved a total of 14 people, 10 clinicians and four scientists, and included 330 publications. It should be noted that there has been a fair amount of collaboration between the members of these committees and about 10 per cent of the papers considered were co-authored by two or more of them. Their research interests are summarised in Table 4.1. In the main, their research has focussed on:

- Molecular biology of lipid pathways and the processes involved in atherosclerosis
- Treatment and management of lipid disorders, CHD and diabetes
- Effect of dietary fats and modifications in diet on lipid pathways and cardiovascular (CV) risks
- Molecular biology, genetics, epidemiology, treatments and management of specific hereditary lipid conditions, particularly FH and familial combined hyperlipidaemia (FCH).



**Table 4.1: Publications of members of the Simon Broome Register group (SB) and Research Committee (R), 2000 onwards.**

Name (committee)	Discipline	Papers	Main research focus
John Betteridge (SB)	Clinician, Endocrinology & Metabolism	43	<ul style="list-style-type: none"> <li>• Molecular biology/ treatment studies concerning lipid pathways and CV function, particularly in diabetes.</li> </ul>
Nigel Capps (SB & R)	Clinician, chemical pathologist	9	<ul style="list-style-type: none"> <li>• Dietary fat &amp; effect of modifications on CVD risk and prevention.</li> <li>• Laboratory methods for analysing variety of substances.</li> </ul>
Muriel Caslake (R)	Scientist, vascular biochemistry	33	<ul style="list-style-type: none"> <li>• Molecular biology/biochemistry of lipid pathways, especially related to atherosclerosis, Type 2 Diabetes, effects of statins, kidney disease</li> <li>• Biomarkers of inflammation as predictors of CHD or of diabetes</li> <li>• molecular biology of FCH and FH</li> </ul>
Paul Durrington (SB & R)	Clinician, lipidologist	55	<ul style="list-style-type: none"> <li>• Role and activity of paraoxonase (PON) in CHD and diabetes</li> <li>• Treatment of hyperlipidaemia in type 2 diabetes</li> <li>• Effect of dietary fats on lipid metabolism &amp; CHD</li> <li>• FH epidemiology, management and treatment</li> </ul>
Bruce Griffin (R)	Scientist, nutritional metabolism	17	<ul style="list-style-type: none"> <li>• Effect of different sorts of dietary fats (fish oils, linoleic acid, polyunsaturated fatty acids) on lipid profile, markers of CV risk and on CVD</li> </ul>
Steve Humphries (SB)	Scientist, cardiovascular genetics	45 <sup>10</sup>	<ul style="list-style-type: none"> <li>• gene variations of lipid, coagulation and inflammation pathways and relationship to clinical measures, atherosclerosis and CVD.</li> <li>• gene-gene or gene-smoking interactions concerning insulin resistance, triglycerides levels or CHD risk</li> </ul>
Rossi Naoumova (SB)	Clinician, cardiovascular & metabolic disorders	22	<ul style="list-style-type: none"> <li>• Non-lipid effects of statins</li> <li>• Genetic studies &amp; management of hereditary lipid disorders, particularly FCH and FH.</li> </ul>
Andrew Neil (SB & R)	Clinician, clinical epidemiology	38	<ul style="list-style-type: none"> <li>• Epidemiology, management &amp; treatment of FH</li> <li>• Treatment &amp; management of diabetes (Types 1 and 2)</li> <li>• Dietary interventions concerning fruit &amp; vegetables and plant sterols</li> </ul>
John Reckless (R)	Clinician, endocrinologist	13	<ul style="list-style-type: none"> <li>• Effects of and tolerance to different treatments for hyperlipidaemia and type 2 diabetes</li> </ul>
Alan Rees (R)	Clinician, endocrinology, diabetes & metabolism	12	<ul style="list-style-type: none"> <li>• Effect of treatments on dyslipidaemia and vascular function, particularly in diabetes.</li> </ul>

<sup>10</sup> Papers for the years 2000 and 2005 only included, see footnote 6, page 99 for further details.

<b>Name (committee)</b>	<b>Discipline</b>	<b>Papers</b>	<b>Main research focus</b>
Tim Reynolds (R)	Clinician, chemical pathologist	46	<ul style="list-style-type: none"> <li>• Discussion and evaluation of different ways of undertaking risk estimation for CHD</li> <li>• Laboratory methods for detecting or monitoring a variety of conditions</li> <li>• The organisation and activities of clinical pathology services</li> <li>• Downs Syndrome screening – ethical concerns</li> <li>• Evaluation of grading systems and equipment for pressure sores</li> </ul>
Mary Seed (SB)	Clinician, CV medicine	11	<ul style="list-style-type: none"> <li>• HRT and lipid management in menopausal women</li> <li>• Different treatments for lipid disorders including drugs and plant sterols/stanol esters.</li> </ul>
Anne Soutar (R)	Scientist, molecular genetics	16	<ul style="list-style-type: none"> <li>• Genetics and molecular biology of FH</li> <li>• Molecular biology of lipid pathways</li> </ul>
Gil Thompson (R)	Clinician, lipidology	24	<ul style="list-style-type: none"> <li>• Lipid and non-lipid effects of statins</li> <li>• Effect of dietary plant sterol and stanol esters in addition to statin therapy</li> <li>• Treatment and management of FH, particularly homozygous FH</li> <li>• Methods for detecting/visualising extent of atherosclerosis and utility in CHD risk assessment.</li> </ul>

In terms of research relating to genetic differences between people, Steve Humphries work is almost exclusively concerned with the influence of genetic variations in cardiovascular disease (CVD). Apart from his work, however, there was only a smattering of research or review papers that included analysis or discussion of genetic polymorphisms related to CV risks or response to treatment. These were mainly co-authored by Caslake (Caslake & Packard, 2004; Freeman et al., 2003), Griffin (Minihane et al., 2000a,b; Paschos et al., 2005) and Durrington (Durrington et al., 2001; Mackness et al., 2000, 2001, 2002a, b, 2004).

The several papers co-authored by Durrington should not give the impression that he particularly emphasises the influence of polymorphisms in his work on CHD. These references include two research papers that include the analysis

of polymorphisms in paraoxonase (an anti-oxidant enzyme found on HDL) and four papers that review the role of paraoxinase in general. These reviews and his own research, in fact, play down the relative influence of genetic variations in paraoxonase. The point is, however, less that his results and his reviews do not support a major role for genetic variations in this enzyme and more that this genetic analysis is included and discussed. That is to say, this illustrates that polymorphism analysis is seen as a research strategy worth undertaking. It should be noted that, in total, these papers still only represent a very small proportion of Caslake, Griffin, Durrington and the other researchers' outputs. Overall, the committee members' main connection with genetics was through an interest in the aetiology and management of specific hereditary lipid disorders.

Research presented at the HEART UK conferences in 2003 and 2004 reinforces this picture. Papers were overwhelmingly concerned with the molecular pathways and management of CHD and lipid disorders and associated conditions including type 2 diabetes, obesity and metabolic syndrome. A small proportion of the papers were concerned with FH and other familial disorders and only one out of about 30 papers presented each year related to genetic polymorphisms. Therefore, although there was a limited amount of interest in genetic variations in relation to CHD in general, this does not represent one of the main aspects of their research.

In terms of the other emerging risk factors for CHD enumerated by Beaglehole and Magnus, the measurement of various biomarkers of the processes of

atherosclerosis was part and parcel of much of the molecular research undertaken by HEART UK members. There was a certain amount of interest in identifying and evaluating new biomarkers of CV risk, including additional lipoproteins or lipoprotein components (Seed et al., 2001; Sweetnam et al., 2000), markers of inflammation (Caslake et al., 2000; Packard et al., 2000) and oxidative stress/anti-oxidant activity (Mackness et al., 2004; Nourooz-Zadeh et al., 2005). The committee members made a minimal contribution concerning other emerging risk factors. Out of the 330 publications considered, just one paper related specifically to infection as a risk factor (Grahame-Clarke et al., 2003), two related specifically to the influence of the early environment (Huxley & Neil, 2004; Rodie et al., 2004) and there was no research relating to the psychosocial environment.

Overall, the contribution of these members of HEART UK to aetiological models of CHD relate mainly to the molecular processes involved in atherosclerosis, focussing on lipid pathways, inflammation and thrombosis. Research related to other emerging risk factors such as the influence of gene variations, infection and the early environment is not a major component of the work of the committee members. However, the abstracts indicate a certain amount of awareness of, or interest in, these areas. This suggests the different elements of CHD aetiology do not represent discrete, but overlapping domains of research. Most importantly, this analysis of the research interests of these HEART UK members illustrates that here, genetic difference is not the dominant discourse concerning CHD.

#### 4.5 HOW TO DEFINE FH?

This section discusses how FH is defined, based particularly on the publications of the Simon Broome register group (Humphries et al., 2005; Huxley et al., 2003; Neil et al., 2003, 2004, 2005; Scientific Steering Committee on Behalf of the Simon Broome Register Group, 1991, 1999). This group is particularly significant in the UK because it introduced the standard criteria for diagnosing FH. The key issue in defining FH seems to be how much this is seen as a clinical syndrome based on a dominant pattern of inheritance of raised cholesterol or of high risk of early CHD, and how much it is seen as a diagnostic category based on the presence of specific gene mutations. This is basically a question about the relationship between FH genotypes<sup>11</sup> and a phenotype<sup>12</sup> of raised cholesterol and of CHD. In the publications of the Simon Broome Register Group, this issue has been played out in two key and related themes:

1. FH diagnostic categories and DNA-based testing.
2. the differential susceptibility to and possible risk factors for CHD for people with FH

These have been discussed against a background of increasing complexity in the genes involved with FH, which is discussed briefly first.

The issues discussed in this section relate very clearly to the central tenets of the geneticisation thesis, in particular:

- the degree to which genes are thought to determine health outcomes

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<sup>11</sup> Genotype is used to describe the specific genetic make-up of a person or of a particular gene variant.

<sup>12</sup> Phenotype means the observable physical characteristics or measurable physiological or biochemical characteristics of a person or a disease, for example eye colour, CHD or hypercholesterolaemia.

- the degree to which people are stratified or distinguished on the basis of their genes
- the adoption of DNA-based technologies to replace other diagnostic techniques
- the degree to which geneticists are involved with or control the identification and classification of FH

### **Genes involved with FH**

In the earliest Simon Broome paper (Scientific Steering Committee on Behalf of the Simon Broome Register Group, 1991: 893), FH was characterised as a dominantly inherited condition of raised cholesterol caused by LDL receptor mutations. A very large number of different LDL receptor mutations have been identified, currently standing at more than 800 (Neil et al., 2005). In the meantime, it has also become apparent that FH can be associated with mutations in other genes. The Simon Broome group first mention this in 2003 (Huxley et al., 2003), discussing the category of familial defective apolipoprotein B-100 (FDAB). This disorder had been diagnosed in the late 1980s (Austin et al., 2004) and it is understood that a small proportion of people with FDAB have the clinical signs and symptoms of FH, accounting for about 4% of cases of FH (Durrington, 2003). By 2005 a third gene, PCSK9, had been identified (Humphries et al., 2005). Therefore, it is now understood that single-gene mutations in a number of different genes can lead to an identical clinical phenotype. To complicate things further, an autosomal **recessive** form of FH has been identified (Durrington, 2003; Soutar & Naoumova, 2004). This rare condition is known as autosomal recessive

hypercholesterolaemia (ARH) and relates to mutations in a different gene. It is clinically very similar to the rarer and more serious homozygous FH.

This genetic heterogeneity necessitated the introduction of new terms to refer to FH. Durrington (2003), for example, talks of the 'FH syndrome', 'clinical FH' and the 'FH phenotype'. FH is no longer defined as a specific single gene defect, but a phenotype resulting from one of a number of (four or possibly more) genes which affect the same molecular pathways. This clearly loosens the link between a single gene and a particular disease, moving away from the one-gene-one-disease monogenic model, to the possibility of a different-genes-same-disease model. This is just one way in which understandings of the genetic aspects of FH have become more complex over the last 15 years. This complexity has emerged through the increasing volume of research on the genetics, molecular biology and clinical outcomes of FH, some of which are addressed in the following sections.

### **DNA-based diagnosis**

By the early 1990s, the molecular mechanisms of LDL metabolism and the genetic basis of FH were well established (Brown & Goldstein, 1986). These were, however, not very important for the diagnosis of FH, which was based on clinical and biochemical signs. The Simon Broome Register Group was instrumental in the UK in establishing a set of clinical diagnostic criteria for FH, introducing the two categories of definite and possible FH. Each was characterised by raised cholesterol, but the presence of tendon xanthomas was seen as decisive. Xanthomas, also referred to as xanthomata, are deposits of

cholesterol on the tendons which occur almost exclusively in FH (Durrington, 2003; Durrington & Sniderman, 2002). Thus definite FH was defined through raised cholesterol and the presence of tendon xanthomas, and possible FH was defined through the presence of raised cholesterol and a family history of either raised cholesterol or early heart disease (see Figure 4.1)

**Figure 4.1: Simon Broome Diagnostic Criteria**

<p><b>Definite FH:</b></p> <ul style="list-style-type: none"><li>a) total cholesterol concentration (in adults +16 years) &gt; 7.5 mmol/l or LDL cholesterol concentration &gt; 4.9 mmol/l AND</li><li>b) tendon xanthomas in the patient or first or second degree relative (parents, siblings, children, grandparents, uncle or aunt).</li></ul> <p><b>Possible FH:</b></p> <ul style="list-style-type: none"><li>a) High cholesterol as defined in (a) above AND</li><li>b) Family history of myocardial infarction before 50 yrs in a second degree relative or before 60 yrs in first degree relative OR family history of raised cholesterol levels &gt; 7.5 mmol/l in first or second degree relative.</li></ul>
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(Neil et al., 2003)

In 2003, Simon Broome publications (Huxley et al., 2003; Neil et al., 2003) start to talk up the possibility of DNA diagnostics for FH. Huxley et al. (2003: 23) introduce the amended Simon Broome criteria, which now include DNA diagnostics. Definite FH is now defined by cholesterol levels in conjunction with the presence of tendon xanthomas and/or ‘DNA-based evidence of an LDL receptor mutation or familial defective apolipoprotein B-100’. It is notable that DNA analysis is an optional part of the diagnosis; the definition remains largely clinical. Huxley et al. (2003) and Neil et al. (2003) introduce a new argument concerning dissatisfaction with the purely clinical diagnostic criteria, based partly on their dependence on the correct identification of tendon xanthomas for definite diagnosis and the ‘the less specific clinical diagnostic



criteria' of possible FH in the absence of xanthomas (Neil et al., 2003: 77).

The papers problematise the possibility of misclassification of FH based on clinical diagnosis alone. DNA analysis is framed as the arbiter of diagnostic decisions:

'There are no entirely satisfactory diagnostic criteria for FH. Clinical criteria...result in false negative diagnosis in 10-20% of relatives of TX+ [xanthoma present] FH cases. By contrast, DNA testing offers a definitive, highly specific, diagnosis, although its sensitivity remains limited. DNA-based criteria are likely to replace clinical criteria, but will not be adopted in routine clinical practice until technical advances have increased the speed and sensitivity of mutation testing, and decreased its cost. In the interim, clinical diagnostic criteria will continue to be used in routine practice'  
(Neil et al., 2003: 77).

The message here is that it is only a matter of time before DNA diagnostics are introduced into routine practice. The current obstacles to its introduction are framed as entirely technical, and temporary. However, one could imagine other possible barriers to DNA based testing concerning for example conceptual, practical, or professional issues. The message of Huxley et al. (2003) is slightly more cautious, raising some questions about the relationship between an FH genotype and FH phenotype. They argue:

'We used the diagnostic criteria of the Simon Broome Register, which place more emphasis on clinical than on DNA-based criteria. The reason is that a small number of LDL receptor mutations appear not to be associated with elevated cholesterol concentrations, and conversely, receptor mutations cannot be identified in all patients with xanthomatous (i.e., definite) familial hypercholesterolaemia'  
(ibid: 24).

In other words, some 'mutations' do not seem to be pathogenic and they cannot find mutations in all people with definite FH. This issue does not appear to have lead to a reappraisal of the overall causal model. The inability to find mutations in patients with definite FH is largely framed as a technical matter:

‘It is not clear whether differences in detection rates relate to genetic heterogeneity of the particular case series, the accuracy of the clinical examination for xanthomata, or differences in DNA methodology’ (ibid, 2003: 24).

In other words it might be attributable to the large number of different mutations in the LDL receptor, or to the limitations of clinical or laboratory techniques. Although caveats are discussed, in the end the paper conveys the same message as Neil et al. (2003), that DNA diagnostics are an inevitable development, contingent only upon technical issues.

The most recent publications, (Neil et al., 2004, 2005), do not use the updated diagnostic categories, they do not discuss DNA diagnosis, nor do they problematise diagnostic categories. However, an abstract from the 2005 HEART UK conference, (Humphries et al., 2005), provides a postscript to this account of the group’s discussion about diagnostic categories, illustrating their continued interest in the area of DNA-testing. It demonstrates that the group is involved in research concerned with the utility of DNA-based testing in UK, based on mutation testing of patients on the Simon Broome register.

Although in the UK, diagnosis remains largely a clinical matter, internationally there has been some move towards DNA-based testing. Most notably, the Netherlands and Norway have introduced national cascade screening programmes that involve genetic tests (Hadfield & Humphries, 2005). The publications of the Simon Broome Group give the impression that it is in favour of DNA-based diagnostic criteria and testing. Yet opinions differ on this area, even amongst the members of the group.

Steve Humphries, member of the Simon Broome group, has been a long-time champion of DNA-based diagnosis for FH (see for example Hadfield & Humphries, 2005; Heath et al., 2001; Humphries et al., 1985, 1997; Marteau et al., 2004). Indeed, Family Heart Association records show that he gave an annual guest lecture entitled ‘Early diagnosis of FH using DNA (gene) probes’ at their AGM in 1987. He is, in fact, a UK expert on FH mutations and custodian of the world-wide LDL receptor mutation database (<http://www.ucl.ac.uk/fh/>), and set up the first DNA diagnostic laboratory for FH in the UK in 1998. His papers have consistently focussed on the benefits of DNA-based diagnosis and the limitations of clinical diagnosis for FH. The benefits discussed are that DNA testing can give an unequivocal diagnosis where cholesterol levels are not decisive, particularly since there is an overlap in cholesterol levels between those with and without FH, and especially useful in children where raised cholesterol levels may develop later:

‘The identification of the FH-causing mutation in a family allows unequivocal diagnosis, and the diagnostic problem caused by the overlap in cholesterol levels between the general population and FH subjects could be eradicated, eliminating false negative diagnosis. A single test, once in the lifetime will be able to ascertain FH status, and early diagnosis in children would also be possible’  
(Marks et al., 2003b: 5).

In this construction, FH status is defined as the presence of a mutation rather than necessarily the presence of raised cholesterol, or high risk of CHD; genotype is the paramount and defining factor, not phenotype.

Humphries’ work acknowledges that there is uncertainty about the relationship between genotype, raised cholesterol and risk of CHD in some cases, talking of cases where people have known mutations, but normal cholesterol levels. It

also acknowledges the difficulty of DNA-based testing in countries like the UK, where there is ‘mutational heterogeneity’. His own work in the UK (Heath et al., 2001), for example, found relatively low mutation detection rates of 32% for adults with definite FH and 14% for adults with possible FH. Like the Simon Broome papers, this is attributed to both technical problems of DNA testing and clinical diagnosis, and aetiological issues concerning the possibility of as yet unidentified further genes involved in FH.

Nevertheless, Humphries and his co-authors still tend to present low mutation detection rates as a mainly technical rather than conceptual issue. Hadfield & Humphries (2005: 430), for example, argue:

‘It is unclear to what extent the low detection rates are due to technical inadequacies of DNA testing (i.e. low sensitivity or ‘false negatives’) or to incorrect clinical diagnosis (i.e. low specificity of clinical diagnosis)’

This implies that if a mutation cannot be found then there is either a problem with the DNA testing or the person does not really have FH, in effect prioritising DNA status over clinical status. In the end they conclude that:

‘It is likely...that for optimal results both genotypic and phenotypic diagnosis should be used’  
(ibid: 430).

Paul Durrington (Durrington, 2001, 2003; Durrington & Sniderman, 2002), a member of the Simon Broome group and chair of the HEART UK research committee, is less positive about DNA-based diagnosis. He again calls into question the relationship between genotype and phenotype, and frames the introduction of DNA diagnostics as controversial and as an issue of priorities:

‘Whether the familial hypercholesterolaemia genotype can be present in people without the clinical syndrome and whether, therefore genetic

testing could be useful is debated...Although the controversy over genetic testing continues, many people with obvious clinical features of familial hypercholesterolaemia who are at risk of premature CHD go undetected or the importance of the diagnosis and the rigour with which this disorder must be treated is not appreciated' (Durrington, 2003: 720).

Durrington prioritises the better recognition of the clinical features of FH. To paraphrase, why bother with DNA testing when so many obvious cases of FH go undiagnosed? This is another example of the 'gap between knowledge and practice' discourse discussed in relation to models of CHD. This may reflect a more general view among lipidologists. However, the subject of DNA-based diagnosis does not appear to have generated much debate among clinicians in the UK. For example, Heath et al. (2001), on the UK molecular testing service, drew no response in the medical journals from UK clinicians. One possibility is that this is just not a burning issue for many clinicians. This argument is supported to a degree by the HEART UK interviews, which will be discussed in the next chapter.

### **CHD risk factors in FH**

Although the Simon Broome publications have discussed diagnostic categories and DNA-based techniques, this is not the focus of their own research. This is the epidemiology and management of FH. The group has produced some of the key research in establishing the mortality rates for CHD and other conditions in treated and untreated FH, demonstrating the efficacy of statin therapy. From the earliest paper, the Simon Broome group was concerned with why some people with FH have very early CHD and others get CHD later or not at all. Their first publication (Scientific Steering Committee on Behalf of the Simon Broome Register Group, 1991) commented that they could not

explain the mortality patterns they had reported, but observed that the age of onset of CHD seemed to run in families.

Since the clinical utility of FH as a diagnostic category is only as a predictor of early CHD, it is not surprising that the Simon Broome group were concerned to work out other factors that might contribute to risk of CHD in patients with FH. The Scientific Steering Committee on Behalf of the Simon Broome Register Group (1999) suggested a number of possible explanations that include both genetic and non-genetic factors. These were that the severity may relate to different mutations in the LDL receptor gene or that response to therapy may differ depending on the mutation, that other genetic polymorphisms may be involved, and the influence of conventional CV risk factors. This shows that the model of CHD in FH is not fixed on the effect of mutations in a single gene, but allows for the influence of other genes and the environment.

The Simon Broome Register Group's own research, to date, has not focussed on differentiating between different LDL receptor mutations, but on other factors. This is demonstrated in Neil et al. (2004: 1431), which argues that people with identical LDL receptor mutations can have different outcomes:

‘Although there is a strong intra-family correlation with the age of coronary death in affected sibling pairs, relatives with identical LDL receptor mutations and similar LDL concentrations may have different outcomes. This suggests that both environmental factors and other genetic polymorphisms influence susceptibility to coronary disease and explain the wide variability in phenotypic expression’.

The paper sets out to test the influence of established and ‘emerging’ risk factors for CHD in people with FH. It is notable that although the excerpt

above mentions the possible influence of other genetic polymorphisms, the research did not include analysis of polymorphisms in other genes and there is little further discussion about such polymorphisms. The research itself included only clinical, that is, phenotypic measures. The paper argues that there is an unequivocal association between the established CHD risk factors with CHD in patients being treated for FH, whereas there was little evidence of an association with the emerging risk factors for this group, concluding that:

‘extensive investigation of risk factors in patients with familial hypercholesterolaemia is not warranted. LDL cholesterol concentration, duration of exposure to raised LDL concentrations, hypertension, and cigarette smoking appear to be the most important modifiable determinants of coronary risk in these patients’  
(ibid: 1435-1436).

This is a clear statement that foregrounds traditional risk factors in the aetiology of CHD in patients being treated for FH, and seems to detract from genetic models. It implies that further work on delimiting particularly severe LDL receptor mutations or the influence of polymorphisms in other genes is unnecessary. Clinical measures, such as LDL cholesterol concentration and how long someone has had raised cholesterol, are seen as more important. This reinforces the clinical rather than genetic orientation in the management of FH.

The group’s focus on traditional ‘lifestyle’ factors is further reinforced in Neil et al. (2005), which reports a significantly lower relative mortality for non-coronary causes in patients on the register, suggesting that this is likely to be due to advice-giving concerning healthy diet, physical activity and smoking cessation. Neil et al. (2004) and (2005) taken together suggest that lifestyle measures, in addition to treatment, contribute to a reduction in mortality from

CHD and other causes in people with FH. The Simon Broome Group's interest in factors contributing to CHD in FH, particularly 'lifestyle' factors, and their lack of focus on the influence of genetic differences suggests a model in which genes are not necessarily the ultimate determinants of ill-health. This is the opposite of the genetic reductionism and determinism anticipated by the geneticisation thesis. The focus on 'lifestyle' factors also reflects the clinical orientation of the Simon Broome Group.

It is notable that none of the Simon Broome analyses include socio-economic class as a variable. This suggests that the group do not consider class to have a bearing on mortality or risk of CHD in people with FH. Furthermore, the 'emerging' risk factors included in Neil et al. (2004) are all biochemical markers of lipid metabolism, inflammation and clotting. In sum, these observations suggest that the Simon Broome Register Group construct the aetiology of CHD mainly in terms of the established risk factors and the processes of atherosclerosis, with a relative lack of interest in the other emerging risk factors or in socio-economic differences in risk. This concurs with the earlier analysis of HEART UK member's constructions of CHD.

Overall, the work of the Simon Broome group has not focussed on the genetic aspects of the aetiology of FH. Their rhetoric of supporting treatment and lifestyle measures is far from a reductionist message that privileges genes. Recommendations urging the continued provision of advice-giving concerning 'lifestyle' measures could be interpreted as focussing on individual level interventions and individualising responsibility as predicted by Lippman.



Nevertheless, clinical indicators of CHD risk are prioritised over genetic indicators.

### **Summary**

To sum up, this section has argued that the Simon Broome group have been influential in the categorisation of FH in the UK and base their diagnostic criteria mainly on clinical indicators. It has suggested that in recent years they have started to problematise these clinical indicators, arguing for a possible role for DNA-based information, and suggested that professionals involved with the Simon Broome group have differing views about the advantages and disadvantages of genetic testing. However, this topic does not seem to have elicited much discussion amongst clinicians more widely in the UK. This section has also discussed the contribution of research undertaken by the Simon Broome group to aetiological models of FH. It has argued that their research does not focus on genetic analysis, and it prioritises traditional risk factors over genetic differences concerning risk of CHD for people with FH

How does this discussion of the aetiology and diagnosis of FH relate to the geneticisation thesis? First, the focus on traditional risk factors and lack of interest in genetic analysis detracts from the genetic reductionism and determinism anticipated by the thesis. Second, although there has been some discussion about DNA-diagnosis in the biomedical literature, FH diagnostic criteria remain largely clinical at the current time in the UK. The increasing stratification along genetic lines suggested by Lippman has yet to materialise in this case. Third, the current picture in the UK, at least, is that DNA

technologies have not replaced other diagnostic techniques. Fourth, the analysis suggests that there may be disagreement about DNA-diagnosis and that this may form along disciplinary lines. Certainly in the UK, the geneticist Steve Humphries has been one of the key proponents of DNA-diagnosis, while there appears to be a lack of enthusiasm or interest amongst clinicians. This is still an evolving area, but at present, it is largely the clinicians not the geneticists who control the identification and classification of FH in the UK. In this case, geneticists do not appear to be the all-powerful group of Lippman's thesis.

#### **4.6 CHAPTER SUMMARY AND DISCUSSION**

The first part of this chapter, based on the analysis of four commentary papers, suggested that there is a genetic vision for understanding, managing and treating CHD that has many of the elements of a geneticised model proposed by Lippman. However, the analysis suggested that there are a number of alternative and competing models of CHD, which draw on a range of emerging risk factors as well as the established ones. Genetic models do not emerge as the dominant idea. The second section demonstrated that the contribution of biomedical professionals involved with HEART UK to models of CHD are largely concerned with the molecular processes involved in atherosclerosis. Little of their research is concerned with the contribution of genetic variations to these processes in general. Their connection to genetics is largely limited to research on specific hereditary conditions, and this research is often concerned with their treatment and management rather than their genetic aspects. Research concerning other emerging risk factors, the early environment,

psychosocial factors and infectious agents, is largely absent. The same model of CHD is supported by the research of the Simon Broome Register Group on variations in CHD in people with FH. This included the traditional risk factors and new markers of atherosclerosis, but not the other emerging risk factors. The papers have largely prioritised traditional risk factors over genetic differences between people with FH.

The chapter has also discussed the definition of FH and the role of genetic testing in its diagnosis. Although diagnostic criteria in the UK remain largely clinical rather than DNA-based, the Simon Broome Register publications have constructed genetic testing as an inevitable development, contingent only upon technical advances. However, research involving genetic testing has raised some questions about the relationship between FH genotypes and a clinical diagnosis of FH and this has led to some ambivalence about the utility of genetic testing, even amongst members of the Simon Broome Register Group. The analysis suggests that different views on this subject may reflect different disciplinary perspectives of lipidologists and geneticists. It is notable that Hedgecoe (2002, 2004b), Kerr (2000) and Shaw (2003) have also raised questions about the clinical utility of genetic tests and/or suggested that there are disciplinary differences between research scientists and clinicians concerning diagnostic categories in relation to several other conditions.

The analysis highlights that there are a number of disciplines with an interest in CHD and it has suggested that constructions of the aetiology and management of this disease might be analysed in terms of boundary work and boundary

disputes (Gieryn, 1983). Boundary work, here, can be thought of as the strategies particular disciplines draw on to demarcate their own discipline from others, and which establish their own expertise, authority and claims on resources in a particular field. In this case, just as Lippman, as an epidemiologist and social scientist, is involved in boundary work concerning geneticists, warning against their colonisation of areas of health and illness which she argues would be better seen in other ways, so the public health physicians and epidemiologists discussed in this chapter were warning against allowing cardiologists jurisdiction over CHD prevention. The discussion of DNA-testing for FH hints at further boundary work between lipidologists and geneticists.

Overall, these data challenge the geneticisation thesis in a number of areas. First, genetics could not be described as the dominant discourse in any of the literatures analysed, apart from those written by geneticists. Second, the Simon Broome Register Group's own research does not focus on genetic analysis and prioritises traditional risk factors in explaining CHD in people with FH. This suggests a model of FH in which genes do not ultimately determine disease. Third, neither the literature on CHD in general, nor the Simon Broome research on FH, focuses on distinguishing people on the basis of their DNA. Fourth, the literature on CHD suggests that the application of genetic technologies in clinical practice remains largely promissory rather than actual. This is also largely the case in the management of FH in the UK, although this differs in other countries. Fifth, the analysis demonstrates that there is a range of biomedical actors involved in the construction of CHD and FH, challenging

Lippman's assertions about the power of geneticists to determine how health problems are defined and managed.

## **CHAPTER 5: HOW DOES HEART UK CONSTRUCT THE FIELD?**

### **5.1 INTRODUCTION**

This chapter focuses on the structure, aims and activities of HEART UK. It first looks at what sort of an organisation HEART UK is in terms of its structure, membership and organisation, in order to consider whether and in what sense it represents a site of lay discourses. It then considers how HEART UK constructs the areas of CHD, hypercholesterolaemia and FH by looking at (1) the aspects of these conditions the organisation prioritises in its activities and membership and (2) the account it provided of the causes of these conditions, paying particular attention to the way genetic elements are presented. The analysis is particularly concerned with the degree to which the organisation's activities and discourses relate specifically to FH and to the genetic elements of CHD, hypercholesterolaemia and FH, and the degree to which it distinguishes between different causes of hypercholesterolaemia.

The chapter is divided into the four main sections: the organisation of HEART UK and the role of patients/lay people; the focus of activities; the aetiology of CHD and hypercholesterolaemia; and constructions of FH. The main sources of data drawn on are the interview accounts provided by senior lay and professional committee members of HEART UK and written material produced by the organisation including information leaflets, the website and annual reports.

The chapter argues that although HEART UK was originally founded by patients, from the accounts provided it is difficult to establish what kind of influence and the degree of influence lay people/patients currently have over the discourses and activities of the organisation. It can be said, however, that it is a highly professionalised organisation. The chapter suggests that the organisation has become less not more focussed on hereditary lipid disorders and that, although an aetiological distinction is made between the hereditary lipid disorders and 'lifestyle induced' hypercholesterolaemia, HEART UK does not stratify patients according to this division in terms of the areas it has become involved in or the support it offers to patients. Finally, it suggests that FH is predominantly framed as a form of heart disease rather than as a genetic disease. These findings place into question a number of the tenets of the geneticisation thesis.

## **5.2 THE ORGANISATION OF HEART UK AND THE ROLE OF PATIENTS/LAY PEOPLE.**

This section is concerned with how the data on HEART UK's constructions of CHD and FH should be understood. It is concerned with the ways in which the organisation's activities and discourses are indicative, specifically, of lay constructions. As discussed in Chapter 2, there has been increasing scholarly interest in patients' associations as sites where expertise may be renegotiated and new forms of knowledge production are emerging (Epstein, 1995; Novas & Rose, 2000; Rabeharisoa, 2003; Rabeharisoa & Callon, 2002; Rapp et al., 2001). My original rationale for studying the Family Heart Association (FHA), the pre-runner to HEART UK, was that it offered a potentially interesting and

important site where disease categories are constructed. Furthermore, it potentially offered a second site of lay constructions of FH and CHD, at a collective rather than individual level. However, as Chapters 1 and 3 discussed, in November 2002 my research plans were overtaken by events. The FHA merged with the British Hyperlipidaemia Association (BHA), the professional organisation for health care practitioners and scientists who specialise in lipid metabolism and lipid disorders, to form HEART UK. This new organisation offered a particularly interesting site for research concerned with the relationship between lay and professional expertise. It raised new research questions about the kinds of relationships that were embodied within this new organisation and the sorts of ideas about expertise upon which the development was founded.

This section provides a brief overview of the history and workings of HEART UK in order to consider whether and in what sense it represents a site where patient discourses are formulated and articulated. The analysis will suggest that although there was some recognition of experiential knowledge, HEART UK maintains a fairly traditional allocation of roles and attribution of expertise. Lay members clearly play an important role in the organisation, but at the present time the discourses and activities of the organisation could not be thought of as strongly influenced by a collective lay discourse in any obvious way. The data on HEART UK should, therefore, be seen as being as much, if not more, about professional as about lay constructions of FH, lipid disorders and CHD.



## **Brief history**

The Family Heart Association (FHA) was founded in its original guise in the early 1980s by patients with hereditary lipid disorders. Its history up to 2002 follows a recognised trajectory (Rabeharisoa, 2003; Wood, 2000), from being run on a voluntary basis to becoming increasingly professionalised through the employment of a small number of professional charity administrators and health care practitioners. By the early 2000s, the FHA had permanent office accommodation, a hand-full of staff, produced a bimonthly newsletter, provided a telephone help-line, and undertook numerous other activities which will be more fully discussed in subsequent sections of this chapter. Although the management of the charity was the responsibility of the predominantly lay committee members, the day-to-day activities were undertaken by the employed staff. Annual reports suggest that there had been some local support groups in the early years, but that these had proved difficult to maintain. The minutes from 1989, for example, report:

‘the groups with a few exceptions, were not working and efforts are being made to restructure these’

and minutes from 1992 report:

‘A number of regional meetings had been held over the year, but attendance did not come up to expectations’.

By the early 2000s, the main gathering of ordinary members was at the annual AGM/members’ day. Annual reports suggest attendance at the AGMs was about 20 people and that total membership was around 1000.

Clinicians were involved with the organisation from the earliest stages. The interview data suggest that they were, for example instrumental in the founding

of the FHA through recruiting some of the early committee members from amongst their patients. There was also a significant amount of cross-over between the FHA and the BHA, with the prime movers in the BHA often also acting as medical advisors to the FHA. Furthermore, AGM minutes report that the nurse-advisors employed by the FHA helped to instigate, and were members of, the BHA's healthcare section, a subgroup specifically for health care practitioners in the field. The AGM minutes also show that latterly, several health care professionals were involved as trustees of the FHA, and that two clinicians went on to become the last two chairs of the organisation before the merge. It is, of course, possible for people to be both clinicians and to have FH, and interviewees talked of one such trustee. The status of the other health care professionals who acted as trustees was not discussed, but it is likely that their involvement was through professional not personal interest. In sum, the FHA, in its later history, could be typified as a health charity **for** people with lipid disorders more than a grass-roots organisation **of** people with lipid disorders.

It is worth digressing briefly to outline the origins and history of a second, connected group, the Simon Broome Heart Research Trust (SBHRT). This was a small charity that was founded by Katherine Broome and Dr Jim Mann in 1977, in memory of Katherine's husband, Simon, who had died of a familial lipid disorder. This charity was important both in the founding of the FHA and in the development of the professional scene. It provided support and funding to initiate the FHA, and Katherine Broome and Jim Mann were involved with the FHA as a trustee and medical advisor, respectively. The charity also

financed meetings for lipidologists at the stage when this discipline was in its infancy, and this grouping of lipidologists went on to develop into the BHA. The charity's main activity was to found the Simon Broome register, the national register of patients diagnosed with familial hyperlipidaemias. The administration of the register was passed on to the BHA, when the SBHRT folded in the late 1990s. As discussed in the previous chapter, the Simon Broome Register Group has played an influential role in the management of FH in the UK. This discussion of the SBHRT has shown that (1) this influential research group was founded through the collaboration of a layperson and a clinician, (2) the charity has played an important part in building the field of lipidology, through helping to initiate the patients' association and the professional's association, and through the research of the register and (3) there have been longstanding links between the SBHRT, the FHA and the BHA.

### **Why merge?**

Interviewees offered a number of reasons for the merge between the FHA and the BHA, but a recurrent theme concerned gaining increased legitimacy with government and other organisations. In part, this was simply a matter of scale; one large organisation rather than two smaller ones could command more fundraising and lobbying clout. As one interviewee commented:

S/HP5: certainly government takes much more notice if you've got five thousand members rather than fifty or five hundred.

It was also concerned with the legitimacy conferred by patient involvement, which must be seen particularly within the context of the current policy climate in the UK of patient and public involvement (see for example Department of Health, 2001b, 2004). This was summed up neatly by one interviewee:

S/HP1: If we are actually to sort of play in the main arena then a lot of, certainly government and a lot of the pharmaceutical industry are much more impressed by the fact that we've actually got patients on the committees than if we didn't have [...] it's more value for money to have a good mix.

Another interviewee commented that professionals' associations might be seen as self-interested, whereas an organisation that included patients would be seen as having more 'altruistic objectives' (S/HP4). The importance of being seen as a group that represents patients is made very clear in HEART UK's Annual Report in 2004, which states plainly that:

'The Charity expects to see greater emphasis upon Patient Membership recruitment and thus a greater voice in the public arena'  
(HEART UK, 2004: 5).

From the patients' perspective, the merge provided their organisation with greater authority, by being associated with some of the leading experts in the lipid field. This was explained by a clinician who had been a member of the FHA:

S/HP2: Well basically what was in it for the patients I think was the perception of some intellectual integrity. You know they, patients' organisations always see themselves as inferior to the doctors [...] They saw it as a way of getting intellectual integrity [...] you know we can say these are all our people, instead of having well these are our medical advisors, which you know were pretty hot-shot people, but actually these people are signed up to our organisation.

This concern with being taken seriously was also evident in the minutes of the FHA's AGMs. In 1999, for example, these talk of the aims of building a strong public image 'as a serious medically authentic organisation' in order to attract funding.

What was notable about the main explanations offered for the merge, was that they were concerned with instrumental or practical reasons. By and large they were not about conceptual ideas concerning collaborative working.

### **Current structure**

HEART UK retained the offices and staff of the FHA, who continued with largely the same functions as before. At the point of the merge, there was a staff of seven full and part-time employees, consisting of the director, the editor of the newsletter, two administrators, two nurse advisors and a dietician. The overall management and direction of the organisation is overseen by the main board and several subcommittees. These are the patient services, the health care, the research, the medical & scientific, and the finance & general committees. HEART UK also took over the administration of the Simon Broome register.

Figures provided to me on my visit to HEART UK in December 2003 were that the organisation had a membership of 1400, made up of approximately 630 'patient' members (lay members) and 770 professional members. Figures provided in the Annual Report in April 2004 differ somewhat, suggesting a membership of around 2500, broken down into about 40 per cent professional members to 60 per cent 'patient' members. Nevertheless, the membership figures are of a similar order of magnitude (low thousands), with a roughly similar division between lay and professional members (similar numbers of each). Within the committee structure of the new organisation, professional members considerably outnumber lay members on the main board of directors

and subcommittees. There were five lay committee members in total, compared with a total of 27 professional committee members. The five lay members all sat on the main board of directors along with ten professional members. These same five lay members also represented lay interest on the subcommittees, with every committee having at least one lay member, with the exception of the Simon Broome committee. They had more involvement in the patients' services committee and in the general & finance committee. There was just one lay member on each of the healthcare, scientific & medical, and research committees. Involvement in these areas was a new role for the lay members and there was a sense that they were still finding their feet. As one interviewee commented: 'I'm still finding out my role in that' (LM2).

The relatively low numbers of lay members involved in the running of HEART UK compared with professional members and staff gives only a limited indication of the position of lay discourses within the organisation. It would be entirely possible for these five lay members to be very influential figures or for the organisation to be highly responsive to the ordinary lay membership. It is, therefore, necessary to provide some details of the relationship between lay and professional members and the perceived role of patients within the organisation.

### **The role of patients in HEART UK**

Interviewees described two major roles for patient committee members. First, they were said to provide valuable input on patient experiences and priorities. It was suggested that it was important to have a pragmatic or real life

perspective on the committees and also at public events. This view suggests a role for experiential knowledge, a knowledge borne from *living day-to day* with the condition, as the following extract suggests:

LM2: we live in the real world, we live with the condition and they don't [...] They're thinking in statistics, in the general population, in their patients, in their research and this is what they're focused on, which is great, but every now and then you have to say hay, you know, it's us you're talking about and this is how we have to cope with it [...] they don't realise what it's like day-to-day living with this.

This was seen as an important part of providing appropriate services for patients and of agenda setting by the organisation, a view expressed succinctly by one of the professional members:

S/PM4: I think it's important to have insights into how the patients feel they are affected by decisions and also to recognise what they see as priorities.

Second, interviewees' talk suggested that patient committee members were valued for their own particular professional skills. Indeed, it was reported that a number of them had been headhunted by the director of the organisation or clinicians in recognition of the useful skills they could contribute. These were people who had business and financial skills, knowledge of charity law, organisational skills and experience of public speaking. This was explained very clearly by one of the lay members:

LM4: Medics are an important profession, but many of them are not brilliant accountants, or lawyers or business men [...] Now patients have all those skills and if the lipid clinics can find them and bring some of them on the committees, you have a much more effective organisation.

The interview schedule did not specifically ask about how the views or experiences of ordinary members, or of other patients, fed into the work of the organisation, and this was rarely discussed in the interviews. As already noted,

there are currently a fairly limited number of occasions when ordinary lay members meet. One interviewee reported that the organisation keeps a record of the questions most frequently asked of the information and support services, and tries to answer these questions in the 'Digest'. This suggests that, in keeping with other patients' associations (Allsop et al., 2004; Baggott et al., 2004), there was at least some formalised way for lay views to feed into the work of the organisation, although the degree of influence is not clear.

In sum, interviewees' accounts suggest that although patient experience (expertise) was valued, to some degree a division of labour was accepted within the organisation, with lay members largely happy to leave scientific and technical matters to the biomedical experts. In this sense, the organisation could be thought of as an 'auxiliary association' (Rabeharisoa, 2003; Rabeharisoa & Callon, 2002), in which responsibility for the production and dissemination of knowledge and practice is delegated to the scientists. It is, however, unusual that these scientists sit in the same organisation.

The current ratio of professional to lay trustees and committee members suggest that the organisation is professionally dominated. The history of staff and clinicians headhunting particularly useful lay members is a further indication of the professionally-led nature of the organisation. The sense that the organisation was and currently remains professionally dominated is reinforced by one interviewee's talk about, and my own observations of, the patients' day and patient workshops, which are the main opportunity for ordinary lay members to meet. There seemed to be a disjunction between the



very technical topics covered and the very practical questions asked by the attendees. The interviewee explained it like this:

LM5: The presentations by medical professionals to the general public were very interesting in their own technical right, but they weren't, dumber down isn't the right word, but simple English, simple analogies weren't there.

Within the organisation, itself, there was a feeling that they had rather low levels of patient involvement and there was talk of scope for greater patient input at the patients' day, and of setting up a support group. This rhetoric of greater involvement is reinforced in the Annual Report of 2004 which states that the directors intend that:

'the place of passive patient [sic] will be replaced with one that sees greater action and involvement in the plans of the Charity'  
(HEART UK, 2004: 6).

It is likely that this rhetoric of greater involvement is being driven by the current policy climate rather than a particular commitment to the notion of lay expertise. Nevertheless, it does demonstrate that HEART UK sees itself as an organisation currently lacking in patient involvement.

### **An FH identity?**

Although HEART UK's future plans include the aim of significantly increasing its patient membership, many of the interviewees talked about the difficulty of retaining members and increasing membership numbers because of the nature of raised cholesterol and FH. In essence, they attributed this to raised cholesterol having no symptoms and to its treatment being unobtrusive. This points to the 'abstract 'disembodied'' nature of the disorder (Lambert & Rose, 1996: 65). They suggested that people tended to seek support or information only at the point of diagnosis and not on a long-term basis. Several illustrated

this by contrasting raised cholesterol with diabetes, which can have symptoms when it is poorly controlled and where treatment may be difficult to manage:

LM1: A feature of the organisation is that people join when they get a diagnosis. After about a year, they don't need you anymore. They feel fine and stabilised. Therefore there's never been a big membership. People manage their ailments and then they don't need help. It's not like diabetes; they're not stabbing themselves.

Another interviewee talked of the difficulty of trying to increase membership numbers because people do not identify raised cholesterol as a disease:

S/HP4: It's largely an asymptomatic disease, which they will see as a condition and possibly not even as a disease, let alone an illness, so it may be much more difficult to get them attuned to this.

The current aims to increase patient numbers would, therefore, seem to involve the creation of a high cholesterol or FH identity that does not presently exist.

While talk of getting more patient members on board can be interpreted as part of the image management of the organisation, reflecting the current involvement agenda of the national government, it was also tied up with normative ideas about what a cholesterol patient should be, in as much as such a person *should* want to be part of a group identified through cholesterol and *should* want to be more informed about cholesterol. This was articulated by one of the interviewees:

LM3: We want to have ten thousand patients on board. Not necessarily on the committee, but subscribed to the magazine, writing to it, the whole background of patients. If you've got bad cholesterol then you should be a member of the society, if only because you'll get the latest thinking and thoughts and opportunities to meet the great and the good, and other patients.

This suggests that HEART UK is not tapping into a ready made constituency, but is actually involved in the construction of such a constituency. This talk

illustrates Rose & Novas' (2004) argument that biosociality can be configured as an obligation, and that this involves a certain kind of biological subjectivity.

### **Summary**

This section has discussed the history and organisation of HEART UK in order to consider the significance of data about this organisation's activities, in other words, what this data can tell us about constructions of FH and CHD. I have argued that:

1. This was already a professionalised organisation before the merge, enjoying the support of clinicians in the lipid field
2. The merge was mainly explained in terms of gaining increased legitimacy and influence, rather than a commitment to collaboration or different forms of expertise
3. There was a recognition of lay knowledge, framed as 'patient experiences', but biomedical matters were largely delegated to the biomedical experts.
4. HEART UK remains professionally-led, with a low proportion of lay members compared with professional members and staff involved in the management of the organisation, and low levels of grass roots activities.
5. The data suggest that there is not a strongly established sense of an FH or cholesterol identity among the patient population that would form the basis of an increase in lay involvement and influence.

In sum, while patients have influenced and continue to influence the direction and activities of HEART UK, it cannot be thought to be influenced by a collective lay discourse in any obvious way. Data about its current activities

and discourses should be seen as reflecting professional constructions of FH, lipids and CHD as much, if not more, than lay constructions.

### **5.3 THE FOCUS OF ACTIVITIES**

This section discusses the overall work of HEART UK and the priorities of the interviewees. It considers the balance of activities between FH and hypercholesterolaemia and CHD in general, and the focus of activities in these areas. The aim is to consider whether the organisation has become more focussed on hereditary conditions and whether it draws a distinction between the different forms of hypercholesterolaemia in terms of the services it offers and activities it undertakes. It will argue that the organisation has increasingly refocused its remit to include cholesterol and CHD risk factors in general, and that interviewees had different views about this expanding role.

#### **Early aims and activities**

The FHA was founded in 1982. Its original title was, in fact, The Familial Hypercholesterolaemia Association. This was amended to the Familial Hypercholesterolaemia and Familial Hyperlipidaemia Association (Family Heart Association) in 1986, and shortened to The Family Heart Association in 1991. The title of the Hyperlipidaemia Education and Research Trust UK (HEART UK) was adopted in 2002 as a result of the merge. Since October 2004 the logo of HEART UK has been appended with the words 'The Cholesterol Charity'. This progression, from the Familial Hypercholesterolaemia Association to the Family Heart Association to HEART UK to HEART UK - The Cholesterol Charity, suggests a decreasing focus on

genetic conditions rather than an increasing focus. The reality is less straightforward.

The minutes of the first AGM, in 1984, state the aims of the organisation as:

1. to make the public and medical profession more aware of this important and fairly common condition [FH].
2. To inform and support those found to have FH.
3. To encourage further research into the cause and treatment of the condition.

These aims focus entirely on FH and are concerned with trying to make sure people with FH were identified and supported. The activities interviewees remembered from these early years focussed on getting publicity for the hereditary disorders, for example by persuading the media to run stories on them. Early activities included producing a diet sheet and a booklet about FH. Although the early aims focussed specifically on gaining awareness of the hereditary lipid disorders, the organisation's activities must be placed within an historical context. It was formed at a time when the cholesterol hypothesis was emerging and was still seen as controversial, and before the introduction of the statins. In some senses, the issue for the FHA was not just to make people aware that raised cholesterol could be an hereditary condition, but that raised cholesterol could be a problem at all. The early activities and comments of interviewees provide a telling reminder about the general milieu concerning eating habits and ideas about CHD at that time in the UK, as one interviewee explained:

LM1: the problem at the time was you couldn't buy low fat products. Skimmed milk was a very rarefied thing. Everyone ate butter, milk puddings, nobody knew about what they should and shouldn't be eating and even if you did it was difficult to find low fat products.

In the absence of really effective cholesterol-lowering medication, it is not surprising that the FHA placed much emphasis on diet. The minutes of the first AGM talks of the organisation being partly responsible for the introduction of new low-fat products:

‘The arrival of new products in the shops such as Tendale and Shape low fat cheeses, Bipro – an egg substitute - and tofu a most useful soybean curd must in part been [sic] influenced by our efforts’.

Although one cannot infer the degree of influence the FHA actually had in the introduction of such products, these minutes are indicative of the kinds of issues that were seen as pertinent at this time. The data illustrate a concern with low-fat diets and low fat products that would have been available not just to people with FH, but to the general public. In this way, the early activities can be seen as contributing to a growing culture concerned with ‘healthy’ or low fat diets in the UK.

Several interviewees talked about a lack of acceptance of lipid disorders. One of the professionals talked about the antagonism of the medical establishment at this time, including the British Heart Foundation, the BMJ, and cardiologists, and the importance of having an organisation that actually recognised FH and cholesterol problems:

S/HP2: British Heart Foundation didn't believe in cholesterol so where were you to get the information and where were you to get support for somebody [...] the BMJ, which we knew as the anti-cholesterol journal, was promulgating people like, very well known people like Michael Oliver, you know, cardiologist, was saying this is a very dangerous thing to lower cholesterol [...] I mean we used to go to meetings and

people would say, God those are the people that run lipid clinics, you know, they've got pointed ears and a forked tail, and we'd say hello cardiologists, don't you wonder how that stuff ever got there in the first place. So you have to remember that [founders of FHA] were working in that environment [...] So they were really up against it.

The controversy surrounding cholesterol and statin therapies at this time has been documented (Lupton & Chapman, 1995; Petersen & Lupton, 1996). This extract also hints at earlier boundary disputes between lipidologists or metabolic specialists and cardiologists, as the field of lipidology was becoming established. A number of interviewees suggested that health practitioners could be ignorant of or dismissive about lipid conditions. LM1 remarked that people would write in asking what they should eat because:

LM1: my doctor says it's rubbish. Most GPs knew nothing about FH and didn't want to know anything about FH [...] people who were going to the GP and saying you know I think I might have this thing were treated more often than not with scepticism by the medical profession.

Part of the work of the FHA, therefore, was to gain increased recognition and acceptance of FH as a diagnostic category, one of the classic aims of lay health advocacy groups outlined by Brown (1995b). LM4 suggests that the organisation had been successful in this aim, contributing to the greater acceptance of hypercholesterolaemia as a disorder:

LM4: there used to be quite a number of GPs who said, oh don't worry about it dear, and don't believe everything you read, and didn't do anything about it. I'm pleased to say that I think partly through the FHA and HEART UK there are a lot less of those about now.

In order to gain increasing acceptance of FH as a diagnostic category it was necessary for such GPs to recognise that raised cholesterol can be problematic in some circumstances. It is, therefore, difficult to separate the early aims and work of the FHA from wider moves to gain increased recognition of lipid disorders and the cholesterol hypothesis in general. The FHA can be thought

of as contributing to establishing the field of lipidology and of establishing cholesterol as a risk factor in CHD, as well gaining wider recognition of the hereditary lipid disorders.

Annual reports suggest that the activities of the organisation branched out from a fairly early stage to include some work on CHD prevention in general, mainly relating to diet and lifestyle. The minutes from 1989, for example, mention the administration of funding for the Family Heart Study. This was one of the main pieces of research that the FHA was involved in and was concerned with cardiovascular (CV) risk factor screening and lifestyle interventions in primary care (see Wood et al., 1994). This broader remit was indicated on the FHA website, whose home page stated:

‘We aim to make all aware that high levels of cholesterol are a serious heart disease risk

We are committed to tracing over 100 000 people in the UK who have inherited high cholesterol

We endeavour to support and advise anyone who wishes to reduce the risks of heart disease’

(<http://www.familyheart.org/pages/topbar.html>, accessed 19/4/2002)

The current aims of the organisation were reported in the first annual report of HEART UK in 2003. This states that:

‘The company was incorporated on 19 July 1991 with the following objectives:

1. To relieve those facing the effects of heart disease and in particular lipid disorders, familial hyperlipidaemias and related conditions.
2. to promote the relief of sickness and the preservation and protection of health by reducing the incidence of vascular disease in the general population of the United Kingdom
3. to provide counselling and advice to relatives of those diagnosed with inherited heart disease



4. to promote the study of causes, diagnosis and treatment of lipid disorders and associated metabolic diseases’.

This shows that the current aims still focus on hereditary lipid disorders, but there is a wider remit which includes the prevention of vascular disease ‘in the general population’ and the promotion of research on all forms of lipid disorders and associated conditions. The wording of this extract also suggests that these extended aims had already been adopted in 1991, long before the merge with the BHA.

### **Current activities**

The main activities of the organisation at the present time can be summarised as: providing information and support to patients; educating and disseminating information to health professionals and scientists and providing their professional forum; lobbying; and representation and advisory work. These activities relate both to hereditary lipid disorders, specifically, and to hypercholesterolaemia and CHD risk more widely, although it is difficult to comment on the balance of work between these areas. The following examples are intended to provide some indication of the different areas of work, rather than an exhaustive account of the organisation’s activities.

Patient information and support services include the telephone, postal and e-mail help-line staffed by health care professionals, the provision of information leaflets and the publication of the Digest, the bimonthly magazine of HEART UK. It was not clear the degree to which queries to the help-line related specifically to hereditary lipid disorders, but the service is publicised in such a way as to attract a variety of enquiries. HEART UK publishes a large number

of information sheets which cover risk factors for CHD, treatments for lipid disorders and numerous leaflets concerned with nutrition and diet. These are listed in figure 5.1. The majority of these leaflets do not relate specifically to hereditary lipid disorders. The Digest magazine carries a mixture of articles on recent research findings and conferences, medications and food products relating to hypercholesterolaemia and CHD in general, with sporadic articles specifically relating to FH.

**Figure 5.1: Leaflets for the general public produced by HEART UK**

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| <p><b>Risk factors and treatments</b></p> <ul style="list-style-type: none"><li>• For your heart's sake: advice for those keen to lower their cholesterol</li><li>• Risk factors for CHD</li><li>• Young at Heart – Heart Disease and the Over 50s</li><li>• Familial Hypercholesterolaemia</li><li>• Children</li><li>• Cholesterol and Lipoproteins</li><li>• Triglycerides</li><li>• Stress</li><li>• Exercise – an Investment in Health</li><li>• Fibrates</li><li>• Resins</li><li>• Statins</li><li>• Glossary</li></ul> <p><b>Diet information sheets</b></p> <ul style="list-style-type: none"><li>• Cooking methods to reduce fat intake</li><li>• Making changes: how to eat less fat, suitable for 'South Asian Diets'</li><li>• What can I do to reduce my fat intake? suitable for South Asian Diets</li><li>• Losing weight</li><li>• Homocysteine &amp; heart disease</li><li>• Guidelines for a Cardioprotective diet</li><li>• Guidelines for a Cardioprotective diet for vegetarians</li><li>• HEART UK diet sheet</li><li>• Nuts and Seeds</li><li>• Oils &amp; fats</li><li>• Fruit and vegetables</li><li>• Fish in the diet</li><li>• Sugars &amp; sweeteners</li><li>• Vitamins</li><li>• Salt</li><li>• Low fat salad dressing</li><li>• Healthy snacks</li><li>• Healthy lunchbox</li><li>• Eating out</li><li>• Eating out best choice</li></ul> |
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Professional activities include the HEART UK annual medical and scientific meeting and Simon Brome register research, discussed in the previous chapter, running study days and producing information and guidelines for health care practitioners. Perhaps the most significant areas of advice concerning professional practice are the Simon Broome diagnostic criteria for FH and

HEART UK's contribution to the Joint British Societies' guidelines on the prevention of cardiovascular disease. These provide guidelines on cardiovascular disease (CVD) risk in the general population and are intended for use in primary care. The FHA and HEART UK have been directly responsible for a relatively small amount of research. This has related to both FH and to lipids in general. The Family Heart Study, mentioned earlier, concerned CHD risks in general practice. More recent research has included projects on family tracking and children's lipid clinics, both concerned specifically with hereditary lipid disorders, and a survey of laboratory lipid services, which related to lipid disorders in general.

Lobbying work again includes a mixture of activities. Certain committee members were instrumental in gaining government funding for a national pilot project on cascade screening for FH, by organising a meeting with the Department of Health. This seems to have been undertaken at the individual initiative of three committee members, rather than through the auspices of HEART UK. Nevertheless, they ensured that the director of HEART UK was present at the meeting. At the same time, HEART UK launched a new joint project with the British Cardiac Patients Association, and some pharmaceutical and food companies, called 'Cholesterol UK', which is a lobbying group concerned with promoting dietary and lifestyle approaches to CV risks and raising awareness about high cholesterol levels in general.

In terms of the representation and advisory work, HEART UK is represented on the steering group of the national cascade screening project for FH and has

submitted evidence to the National Institute of Clinical Excellence (NICE) appraisal on FH. It has also submitted evidence to NICE and other regulatory bodies on issues concerned with CVD in general, including obesity, lipids and CVD, statins, the sale of over the counter statins, and on clopidagrel (an anti-platelet drug used in heart disease).

In sum, the organisation undertakes a range of activities which are concerned with both hereditary lipid disorders specifically and hypercholesterolaemia and CVD in general. It is not possible to assess, from this list of activities, the balance of work between hereditary lipid disorders and CVD risks in general.

### **Interviewees' priorities**

Interviewees' accounts of the aims and activities of HEART UK tended to acknowledge the span of work discussed in the previous section. However, the interviewees emphasised different aspects of this work. S/HP5 talked almost exclusively about cholesterol, lipids and CHD in general, and about associated conditions such as obesity, diabetes and stroke, with little mention of familial disorders. By contrast, S/HP2 focussed entirely on FH. These interviews represent the two extreme cases. Overall, interviewees tended to place familial disorders at the centre of the organisation's work, whilst acknowledging an extended role concerning the problem of high cholesterol in general. In the following extract, for example, S/HP3 describes the aims of HEART UK, suggesting that the primary focus for the organisation is on *improving the lot* of patients with hereditary lipid disorders, which constitutes *the bottom line*:

S/HP3: To provide the fullest possible support service for patients with inherited FH and/or FCH [familial combined hyperlipidaemia]. That's

the primary role. The secondary role is to make the general public aware of the first in order to track more people who inherit the disorder. The support behind that comes in primary and secondary prevention. It is therefore important that they [health care professionals] are also provided with that education and awareness. In addition, it's important to support research wherever possible into improving the lot for patients. That's basically the bottom line. But as we have a problem with raised lipid levels anyway, we should also be engaging the public generally with virtually the same messages. Understanding that between fifty and sixty per cent of the population have a total cholesterol level which is above that which is recommended.

LM4 expressed a similar view, saying that FH is still at the *centre* of the organisation's concerns, but that the organisation should not distinguish between people with FH and other people with raised cholesterol in terms of the services offered to patients:

LM4: We are the cholesterol charity, not necessarily familial

KW: And is that a move that you've gladly seen from the family cholesterol charity to the cholesterol charity?

LM4: Yes, it's not entirely removed, the concentration, the centre of it is still familial hypercholesterolaemia, but there is the inevitable, if you're doing that, the government says, well what about the others? There's only a hundred thousand of these [people with FH]. The lipid clinics, some of them are not familial patients, no family history, no genetic involvement, but they have very high cholesterol, so we can't say we don't want you, you go off to someone else.

The extension of work to include general issues of cholesterol and CHD was associated with a certain amount of ambivalence. When LM4 talked of an inevitable overlap of interests between the familial conditions and CHD in general, he was cautious because this led to a cross over of remit between HEART UK and other heart charities:

LM4: Originally it was concentrated on FH and FCH [familial combined hyperlipidaemia] and other inherited diseases, but there is an inevitable overlap about which we have to be a bit cautious. I would say we are the cholesterol charity now, but we do have to be a little careful not to be setting ourselves up as the heart charity because there are other large heart charities who would be very put out by that.

S/HP4 was more direct about his views on the remit of HEART UK, arguing that it should remain primarily concerned with the familial lipid disorders, and was somewhat critical of the past activities and focus of the FHA:

S/HP4: certainly the FHA under a number of its directors, I think sometimes mistakenly, went way beyond familial hyperlipidaemias and FH in particular and was involved for instance with MSD [pharmaceutical company] and the know your number campaign which was really dealing with issues around polygenic hypercholesterolaemia [general raised cholesterol] and population risk of heart disease [...]. I've no doubt that HEART UK, if invited, make a positive contribution to general issues around lifestyle advice and sorting diet and so on, but I don't see that as being specific to HEART UK. It's done by British Hypertension Society, BHF [British Heart Foundation], British Cardiac Society. What I'm saying is, what is it which is specific and different about HEART UK, and it is that not only is it concerned about hyperlipidaemias in general, but its major remit seems to be, to me, those individuals that have got inherited dislipidaemias, 'cos there's no other group which sees them as its *raison d'être*.

By contrast, LM3 talked of the hereditary lipid disorders as having become a *subset* of the organisation's work because of the general expansion of the construction of cholesterol as a problem:

LM3: It's growing very fast because cholesterol is becoming a big issue and while we started with [hereditary] hyperlipidaemia, I wouldn't say it's become a subsidiary, but it's becoming part of the cholesterol issues and problems, almost a subset within the whole.

## **Summary**

This section has shown that the original aims of the organisation were exclusively concerned with hereditary lipid disorders. The analysis suggests that the remit of the organisation widened to include some work on cholesterol as a risk factor in the general population more than a decade before the merge with the BHA and that current aims and activities cover both hereditary lipid disorders and CVD more generally. However, HEART UK interviewees had a variety of views about the main focus of the organisation and the degree to

which it had or should expand its role away from the hereditary lipid disorders. Overall, the analysis suggests that the organisation has become less focussed on genetics during its history, in as much as it has expanded its remit from being concerned exclusively with hereditary lipid disorders to a wider focus including population-wide CVD risks, prevention and treatment. Furthermore, even the early concern with hereditary lipid disorders must be seen as contributing to establishing and raising the profile of the cholesterol hypothesis in general. In terms of the geneticisation thesis, it appears that, in this example, genetic discourses have in some ways become **less rather than more dominant**, and that HEART UK does not distinguish between the causes of hypercholesterolemia in fulfilling its remit to support and inform patients.

#### **5.4 THE AETIOLOGY OF CHD AND HYPERCHOLESTEROLAEMIA**

This section discusses how HEART UK publications and the HEART UK interviewees present the causes of CHD and of hypercholesterolaemia. It will suggest that, like the biomedical constructions discussed in the previous chapter, there is relatively little interest in polygenic influences in CHD, but a focus on ‘malleable’ or ‘lifestyle’ risk factors, framed as diet, exercise, stress and weight. It will argue that HEART UK’s interest in genetics is mainly concerned with specific hereditary lipid disorders rather than polygenic influences as a more general risk factor for CHD, and that in aetiological terms, a divide is maintained between hereditary lipid disorders and other hypercholesterolaemia.



Information leaflets produced by HEART UK (see figure 5.1) present the cause of CHD as multifactorial and discuss it in terms of risk factors and their additive effects. Risk factors are divided into two types: things that cannot be changed, i.e. age, sex, family history; and things that can be changed through ‘leading a healthy lifestyle’ (see figure 5.2). These ‘modifiable’ risk factors include all the established risk factors, i.e. cholesterol, blood pressure, diet, smoking, obesity, lack of exercise, and also ‘excessive stress’. The inclusion of stress must be seen as a recognition of the role of the psychosocial environment, categorised by Beaglehole & Magnus (2002a) as a new risk factor. HEART UK produce a specific leaflet on homocysteine and heart disease, again a recognition of one of the new biomarkers of CHD risk.

**Figure 5.2: What Causes CHD?**

<p>CHD is more likely:</p> <ul style="list-style-type: none"><li>• With increasing age</li><li>• In men rather than women before menopause</li><li>• If there is a strong family history of CHD</li></ul> <p>These risk factors cannot be changed. But there are others that <b>can</b> be modified by leading a healthy lifestyle. These include:</p> <ul style="list-style-type: none"><li>• Elevated blood cholesterol</li><li>• High triglycerides with low HDL</li><li>• Elevated blood pressure</li><li>• Diabetes</li><li>• Smoking</li><li>• Obesity</li><li>• Inactivity</li><li>• Excessive alcohol</li><li>• Excessive stress</li></ul>
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Taken from: ‘For Your Heart’s Sake: Advice For Those Keen To Lower Their Cholesterol’ (HEART UK, n.d: 3, original emphasis)

The leaflets make very little reference to genetics, apart from sections that relate specifically to FH. They mention that high blood pressure ‘tends to run

in families' but go on to say that it is also influenced by lifestyle. The amount of homocysteine a person produces is also said to be 'influenced by your diet and your genes'. These two references constitute the total discussion of the influence of genes on CHD, in general, within the leaflets. All further references are concerned with specific hereditary conditions. Raised cholesterol is framed as being either due to lifestyle factors, mainly eating the wrong foods, or to an inherited disorder:

'Cholesterol levels may rise due to lifestyle factors, such as eating a diet rich in saturated fat. High cholesterol may also be inherited, as in the disorders of familial hypercholesterolaemia (FH) and familial combined hyperlipidaemia (FCH)'  
(HEART UK, n.d: 3)

In sum, the leaflets are strongly focused on 'lifestyle' or 'modifiable' factors, with most leaflets concerned with diet. The influence of polygenic gene variations on cholesterol levels or other aspects of CHD risk was practically absent. Two classes of hypercholesterolaemia are set up: (1) hereditary and (2) lifestyle induced.

A very similar picture emerged in the interviews with members of HEART UK. There were no unprompted references to the possibility of genetic susceptibility to CHD beyond the well-established hereditary lipid disorders in interviews with either professional or lay members. The only occasion on which the influence of genetic variations on cholesterol levels was mentioned, the issue was introduced by me. The interviewee was discussing why people with FH have variable susceptibility to CHD and was stressing the importance of the established CHD risk factors. I posed a question about the possible influence of other genes:

KW: and there's no suggestion of any multi-gene or polygenic influences or interactions at all?

S/HP4: There certainly isn't in the sense that if you have an LDL receptor mutation, it's not associated with any other genetic abnormality which enhances risk [...] It's simply that anyone with heterozygous FH can be exposed to environmental risk factors the same as anyone else, and they may or may not have the good fortune or bad fortune to have inherited high or low rates of, levels of HDL cholesterol for instance.

This single reference to a genetic influence on HDL levels illustrates that the interviewee recognised a wider role for the influence of genetic differences on cholesterol metabolism. The reason this subject commanded so little of his attention is perhaps because these genetic differences are not seen as modifiable. He went on to emphasise the need for lifestyle advice for FH patients.

Apart from this, HEART UK interviewees framed the causes of hypercholesterolaemia mainly in terms of hereditary cholesterol conditions and lifestyle issues, with occasional references to the influence of aging or menopause. Just as with the information leaflets, the overall effect of interviewees' talk was to construct two classes of raised cholesterol, due either to the established heredity disorders or to lifestyle. As the data on HEART UK's activities have demonstrated, this aetiological distinction does not delimit the interests of HEART UK.

Given the lack of discussion of the possible influence of genetic variations on cholesterol metabolism, it seems strange that both the biomedical literature and the professional members interviewed refer to the ordinary, not specifically hereditary raised cholesterol as 'polygenic hypercholesterolaemia'. This term

implies the involvement of many genes. Durrington (2003:722) explains polygenic hypercholesterolaemia as follows:

‘The main causes are high fat intake, particularly saturated fat, and obesity. Genetic factors are also assumed to be important, because individuals vary in their cholesterol response to diet. However, there is no clear pattern of inheritance; a combination of more than one genetic variant is generally required for this type of hypercholesterolaemia (polygenic inheritance)’.

This definition suggests that people have a variable response to dietary changes and that this is attributed to variations in a number of genes. The idea that cholesterol levels respond differently to dietary changes in different people was discussed by Brett (1991) more than a decade earlier in an effort to dissipate the moral undercurrents of both professional and lay constructions of hypercholesterolaemia. Brett’s arguments contribute to the more general critique of public health discourses concerning CHD prevention (Davison et al., 1989, 1991, 1992; Sachs, 1996; Petersen & Lupton, 1996). These arguments seem to have had little sway here, as they are hardly present in the discourses of HEART UK, and lifestyle remains central to constructions of hypercholesterolaemia. Therefore, although the term polygenic hypercholesterolaemia embodies a genetic aetiology, there was little interest in genetic influences and the term seems to be employed to mean raised cholesterol caused by lifestyle. This was nowhere more clear than in the interview with S/HP2 who referred to ‘polygenic, lifestyle-induced high cholesterol’ and in a paper co-authored by members of HEART UK which referred to ‘non-genetic polygenic hypercholesterolaemia’ (Marks et al., 2003:2). The idea that a condition can be ‘non-genetic’ and ‘polygenic’, in particular, seems contradictory and confusing. The data suggest that, in the case of hypercholesterolaemia, the term polygenic is used in order to make a

clear distinction to monogenic or more clearly hereditary forms of the condition. Polygenic seems to mean a generic form of hypercholesterolaemia, without implying any specific interest in genetic variations.

The model of CHD and hypercholesterolaemia presented by HEART UK continues to imply a high degree of personal responsibility. However, there was rarely an explicit blaming discourse. Indeed, I only recorded one example of this, which occurred at the patients' workshop at the HEART UK conference in 2003. On this occasion, a clinician was answering questions about the use of LDL apheresis, a treatment reserved for people with homozygous FH or where statins are not effective or not tolerated. The clinician commented:

‘Millions of people have got high cholesterol due to their lifestyle. That’s your fault. You brought it on yourself. We’re not interested in them. People with FH can’t help it’

One can only speculate that the use of this particular rhetoric relates to the context in which it occurred. It was an off-the-cuff comment to a lay audience made up predominantly of people with hereditary lipid disorders and their partners. It is likely that the clinician enrolls more neutral language in her written accounts of LDL apheresis and in presentations to other professionals. Nevertheless, this illustrates that it is possible for clinicians to slip into an explicit blaming rhetoric.

HEART UK's focus on 'lifestyle' factors is indicative of its fairly traditional health promotional approach that focuses on encouraging individuals to make behavioural changes. There is little, if any, reference to social inequalities in

the distribution of CHD that have long been a theme in epidemiology and in more recent government policy in the UK (see Davey Smith, 2003; Department of Health, 2000; Marmot & Wilkinson, 2005). There was some recognition of ethnic differences in the distribution of CHD and diabetes, illustrated by the two leaflets that focus specifically on low fat diets suitable for South Asian people, and a small number of research papers by HEART UK members concerned with molecular mechanisms and ethnicity. Direct reference to, or research concerning socio-economic class was absent and there were only a few rather veiled references to socio-economic issues. LM1 talked of healthy eating as a middle-class attribute, which she associated with education:

LM1: my children do eat what you would call more healthily nowadays, but then everybody, well most people with education do on the whole, the chattering classes I should say.

LM4 also talked about getting people to eat a healthy diet, connecting this with both education and economic opportunity:

LM4: it certainly is more expensive to buy only healthy foods still. It's as so often, it's those who can afford it who are the healthiest and those who can't, not only know less about it, but can less afford to be healthy. A shameful situation and I'd like to see something done about it.

This subject was almost entirely absent in the interviews with professionals.

## **Summary**

This section has argued that HEART UK's leaflets and the interviewees construct two classes of hypercholesterolaemia, hereditary lipid disorders and lifestyle-induced raised cholesterol, and the organisation focuses particularly on encouraging individual-level behavioural changes. There is practically no reference to the possible more minor influence of other genetic variations in lipid metabolism or to polygenic influences in CHD. While this construction

of CHD aetiology may individualise responsibility for CHD and detract from wider socio-economic determinants of health, as Lippman has argued, this cannot be attributed to a geneticised discourse about aetiology. This section has argued that polygenic influences are not dominant in HEART UK's public discourses. This is consistent with the biomedical constructions reported in the previous chapter.

## **5.5 CONSTRUCTIONS OF FH**

This section is concerned with how HEART UK constructs FH and the degree to which it is framed as a genetic disease. Following on from the previous chapter, this section will first discuss interviewees' views on DNA diagnosis. It will then discuss in broader terms whether FH was framed in a genetic way through the activities of HEART UK and through interviewees' talk. It will be argued that the interviewees had a variety of views on DNA diagnosis, but that it was largely not seen as a priority. Furthermore, the organisation tended to frame FH as a form of CHD rather than framing it through a genetic lens.

### **DNA diagnosis**

The previous chapter suggested that although the Simon Broome Group publications appeared to support DNA-based testing for FH, its members are not universally enthusiastic about it, and that this area did not seem to have elicited much discussion among clinicians in the UK. In the interviews with the staff, lay and professional members of HEART UK, only one interviewee, S/HP2, raised the issue of DNA-based diagnosis spontaneously, arguing that it was one of the main priorities concerning developments for people with FH. In

the other interviews, the discussion of the topic was prompted by me asking a specific question, or was entirely absent from the interview. This certainly suggests that it is largely not a topic at the forefront of people's minds. When asked, people involved with HEART UK had a mix of views on this topic, which largely aligned with the arguments in the biomedical literature discussed in the previous chapter.

For S/HP2, DNA-testing was a priority area; a genetic diagnosis was seen as giving an unambiguous indicator of high risk:

S/HP2: What would help so much would be to have a decent really sensitive and specific genetic screening tool. Because at the moment we pay nearly five hundred pounds to one of two centres and they tell us, well this person's got this mutation or that mutation, but actually we can only ever pick up fifty per cent of FH patients, so you know, we're still left in the dark as to exactly what the risk is to an individual patient.

Rehearsing an argument found in the literature, she suggested that the particular advantage of DNA testing was to be able to give an unequivocal diagnosis to people whose cholesterol is in the 'middle ground'. Her argument was that it was important to be able to distinguish people with FH who have moderately raised cholesterol, because they have had it from birth and are therefore at greater risk than other people with the same moderately raised levels of cholesterol. This means that getting the right diagnostic label was about disease management, as FH patients, even with only moderately raised cholesterol, warrant much more intensive monitoring and treatment. The issue was to be able to *draw a line* based on genetic analysis:

S/HP2: the biggest problem is around those patients that have a cholesterol of 7.5 and above, because the Simon Broome Register gives possible FH as over 7.5. Now the problem is I have a patient before me who has a cholesterol of 8.2. Now out there are stacks of people with



cholesterol of 8.2. [...] If you were able to have a test, a genetic test, that said oh yeah this man's got FH, you know immediately this person must be on treatment, family history must be given and we must screen him annually, perhaps exercise test or whatever, so that's what it's all about [...] It's not like the 14s, the 15s, the 12s, cos that's obvious. It's that sort of middle ground where potentially patients are really really disadvantaged [...] you really need to be able to draw a line and say this person with a cholesterol of 7.8 has got FH therefore they've had it from birth, therefore the damage has happened from birth.

S/HP4, when asked, was also largely positive about DNA diagnostics, suggesting that it would be particularly useful in diagnosing children, and in adults where either the family history was not available or clinical signs were inconclusive. Other interviewees were more equivocal about it, prioritising clinical features over genetic information. S/HP5, for example, talked of the limitations of genetic testing in terms of the expense, the large number of mutations and the inability to find mutations for a large minority of FH patients. His main point was that high risk of CHD was the important aspect, regardless of genetic status. The category of FH was not the paramount concern, but the clinical aspects of high cholesterol and early CHD that runs in families, whether or not this was labelled FH. This is illustrated in the excerpt below, which suggests that the interviewee is not antagonistic towards DNA testing, he is just not excited about it:

S/HP5: In practice you do it on family history, age and using the criteria under the Simon Broome Trust. That is probably clinically the best way of doing it. Because at the end of the day what you want to know is what their risk is. It may not be FH, but if it's got a familial component and they get their coronaries early, they need treating anyway. So on a purely pragmatic point of view that's probably the way to go. Yes, fine, by all means we've no objection to organising what's been dubbed a DNA bank and that they're tested and checked. That isn't an essential part of the cascade screening system. And I would go with either.

The discussion of DNA testing in the previous chapter hinted that this could be seen as a boundary issue concerning the role of geneticists and clinicians. The wide introduction of DNA testing in the UK would see a shift in professional practice. It would move the management of FH from the current situation, in which FH is practically the sole domain of clinicians, to a situation in which geneticists and the genetic services might have increasing influence. The fact that some health care practitioners were positive about DNA diagnosis and others were less enthusiastic suggests a more complex picture, with differences of opinion not aligning strictly on disciplinary lines. Nevertheless, the idea of a boundary dispute between clinicians and geneticists is also hinted at by data from one of the interviews:

KW: Do you have a view about whether we should be pursuing DNA-based diagnostics or whether it should be on the clinical signs?

S/HP1: I think that's a little nugget that comes up at every conference that we run. You know you have the geneticists on one side and clinicians on the other. The clinicians are louder than the geneticists.

The interviewee frames DNA research as a rather esoteric activity, and is hard-pressed to understand the clinical relevance of DNA-testing, gently making fun of the scientists who are involved in mutation identification. The jocular way the interviewee talks about this area and her focus on the 'clinical significance' reinforces the idea of a divide between scientists and health care practitioners on this issue. Like the previous interviewee, her focus is on the clinical indication of high cholesterol, which is seen as more important:

S/HP1: at the end of the day, you have high cholesterol, you treat it with a statin, but you don't necessarily need to know what particular mutation they've got. There are so many different mutations [...] until somebody can actually perfect it [gene therapy] I really don't know that we need to actually pursue the DNA. It's a bit like stamp collecting to be honest. You sit in a room with all these people who've worked out these different mutations and you sort of say well big deal, you know, it's x13457 instead of x13456 fantastic, and you sort of say, what is the

clinical significance of that? And they all go, but it's x13---, it's a blue stamp instead of a pink stamp, and you just think well, clinically that won't mean a lot to a patient. They will not care what sort of LDL mutation they have got. What they are worried about is have they passed it onto their kids and can you treat it.

Lay members were similarly disinterested in DNA-diagnosis. LM2, like S/HP5 was less fixed on the specific diagnostic category of FH. In fact she did not distinguish between inherited forms of raised cholesterol and other lipid disorders. In her view the important factor was raised cholesterol. As a result DNA-based diagnosis was a low priority:

KW: do you see any benefits to a DNA-based test?

LM2: No because if you've got raised cholesterol, whether it's inherited or not, you've got a problem that if it's not treated you're going to have a heart attack or a stroke, so to me it doesn't matter, that's my personal view [...] Even today people don't get genetic tests. I mean this is a genetic condition, but you don't get a genetic test. If your cholesterol's raised and you've got a family history like mine then the likelihood is you've got FH. You don't need to do a genetic test to treat people and even if it's not FH, if you've got raised cholesterol you know, you need help, so.

Two other lay members were not aware of DNA-testing as a current debate. It was not a subject that they had particularly engaged with. As LM4 said 'It's not something I've given much thought'. In sum, DNA testing was not at the forefront of HEART UK's agenda. There was mainly a lack of support or lack of interest in DNA diagnostics among these key figures in HEART UK. This could not necessarily have been anticipated on the basis of the arguments presented in the Simon Broome publications, discussed in the previous chapter.

### **Genetic discourses?**

Although a large part of HEART UK's work focuses on the familial lipid disorders, there was very little within the organisation's structure or activities, or interviewees' talk that framed these in a genetic way. The organisation's

professional trustees and committee members are mainly specialists in metabolism, lipids, cardiovascular disease or chemical pathology, and include just two molecular geneticists and no clinical geneticists. HEART UK is represented on the steering committee of the London Genetics Knowledge Park and on the Genetics and Insurance Committee, a government advisory body. However, these were the only organisations mentioned that are specifically focussed on genetics. It is notable that collaborative working concerned with education, lobbying, and patient representation is largely oriented towards CVD rather than genetic disease, and a large number of organisations were mentioned in this respect including Diabetes UK, the Primary Care Cardiovascular Society, the Joint Federation of Primary Care Societies, the British Cardiac Patients Association, The British Cardiac Society and the British Hypertension Society. These are organisations, for example, with whom HEART UK organises conferences or study days for health care practitioners. This orientation towards CVD was also illustrated very clearly in the interview with S/HP3. When asked about the types of groups HEART UK collaborates with, his immediate response was to talk about the ‘problem’ of CHD in general. When asked directly about membership of the Genetics Interest Group (GIG), the national umbrella group for patients’ organisations associated with genetic disorders, he explained that HEART UK is not a member *because you can’t do everything*:

KW: Who are the main sorts of groups that you collaborate with?

S/HP3: You have to seek partnership. What we are dealing with is a major problem. Heart disease is right there at the very top of the list. It kills more than anybody else’s disease or disorders so we’ve gotta do something serious about it [...] we actually have extremely good relationships with small organisations who are in the heart charity business. We should be much stronger within the BHF [British Heart Foundation] and we constantly work on that. We have good relations

with Diabetes UK, we have good relationships with important people working in the field cardiology and within the commercial sector [...]

KW: What about the genetics umbrella group, GIG?

S/HP3: I think by default we're connected with them. I sit on a sort of committee, which is called the Genetics Knowledge Park in London, Alistair Kent [Director of GIG] also sits on the same committee, but as I say you can't do everything.

In the UK context, GIG can be thought of as a defining group concerning genetic conditions. This interviewee's comment provides a strong indication of where HEART UK positions itself conceptually and politically. His talk had the effect of framing FH as a subset of CHD and suggests that specifically genetic related activities are not prioritised. This reinforces the construction of the hereditary lipid disorders as being more to do with heart disease than with genetic disease.

A comment by S/HP1 further reinforces the idea that FH is not predominantly constructed within a genetic frame. This comment refers to the national cascade screening project for FH, which was introduced by the genetics White Paper (Cm 5791 - II, 2003). To understand this comment, it is important to note that the screening project will be based on clinical diagnostic criteria, although there was some talk of seeking extra funding to attach some additional research on DNA-diagnosis to the main pilot:

S/HP1: Heart UK has been part of the battle to get money out of the genetics White Paper. On genetic screening and cascade screening [...] Money's come through the genetics people, that isn't for genetics. They've got to get their money for the genetics bit separately.

This talk of money for cascade screening coming through the genetics White Paper, but not being *for genetics* and of having to get the money *for the*

*genetics bit separately* frames genetics as something specifically to do with genetic testing, rather than genetic disease or hereditary disease.

One area where there was explicit reference to genetics was talk about possibilities for the future. Lay members talked of their hopes that one day there would be direct treatment of the genetic causes or of gene replacement therapy for FH. This was discussed in quite a speculative way, perhaps because it was an area that they did not feel very competent to comment on:

LM4: I think in the medium to long term future the fundamental causes, the genetic causes of the familial conditions are very important, in our core concern with them. Genes have been identified, and again it's no good asking me the details, and maybe in the future direct treatment of the genetic cause will be possible.

Nevertheless, the topic was not a large part of any interview or portrayed as a major priority. This subject was absent from the research and interests of professional members.

There was one notable exception, where an interviewee did frame FH in a genetic way. S/HP2 talked about the possibility of genetic counselling and reproductive decision-making in relation to FH, drawing on arguments about *individual choice* and people having *options*. She argued that these options were contingent upon the availability of DNA-testing:

S/HP2: You would definitely advise about family screening, you'd be able to give genetic counselling to people thinking about having children you know. You've got a one in two chance of it having, of a baby having the condition. So all of that comes down from knowing that [...] Then I think in discussion you have to be able to offer patients that option, like you know it's not like Huntington's Chorea but I mean [...] patients may choose not to know but I think we have to have the option to offer them that because it's their life and they need to have that option.

These arguments about the provision of genetic counselling and the possibility of reproductive decision-making for people with heterozygous FH were unique to S/HP2. They were not present in any other situation, neither in interviews with HEART UK members, nor in the biomedical literature nor at the conferences.

### **Summary**

To sum up, HEART UK interviewees have different views on DNA-testing, but they tend not to see this as a priority. Following on from the discussion in the first chapter, the data support the idea of a boundary dispute between clinicians and geneticists on this subject, at the same time as complicating this argument. The analysis raises questions about how much use clinicians would make of genetic testing for FH in practice. FH was predominantly framed as a subset of CHD, rather than as a genetic disease. Cox and Starzomski (2003: 162) have described a similar lack of focus on genetics in the construction of polycystic kidney disease (PKD) arguing that it is defined through ‘a kidney disease rather than a PKD culture’. In a similar way, HEART UK’s structure and activities in relation to FH could perhaps be described as being infused with a CHD culture rather than a genetic culture. There was, however, one notable exception, which illustrates that it is possible to see FH through a genetic lens, linked with talk of life plans and choices. In terms of Lippman’s thesis, this analysis again suggests that (1) genetic discourses are not dominant. Indeed, they are barely evident at all. (2) Heart UK interviewees were not, in the main, interested in distinguishing people on the basis of DNA. They tended to privilege clinical status instead. (3) HEART UK was largely

uninterested in the potential application of genetic technologies in the diagnosis and management of FH.

## **5.6 CHAPTER SUMMARY AND DISCUSSION**

This chapter has focused on the aims and activities of HEART UK. It has suggested that lay people were instrumental in founding the FHA and the Simon Broome Register and continued to influence the direction and activities of the FHA/HEART UK. However, the organisation became increasingly professionalised and currently appears to have low levels of lay influence. Its current discourses and activities must, therefore, be seen as an expression of professional constructions as much as, if not more than, lay constructions of CHD and FH. The analysis shows that the original focus of the FHA was solely on hereditary lipid disorders, but broadened out from an early stage to include CVD risks in general. Nevertheless, even from the earliest times, the organisation contributed to establishing the cholesterol hypothesis and the field of lipidology. While HEART UK may distinguish in aetiological terms between hereditary lipid disorders and ‘lifestyle induced’ hypercholesterolaemia, the organisation does not make such distinctions in terms of the support it provides to patients. Overall, there was very little discussion of genetics in relation to either CHD risks in general or in relation to FH, and FH was predominantly framed as a form of heart disease rather than as a genetic disease. Relating these findings to the geneticisation thesis, the analysis suggests that:

1. Genetic discourses are certainly not dominant within HEART UK. In fact, there has been a move away from genetic discourses, to some



degree, as the remit broadened to include cholesterol and CHD risks in general. Genetic constructions of CHD in general were barely evident, and there was very little specifically genetic talk concerning FH.

2. Heart UK does not distinguish between patients on the basis of their DNA, largely privileging clinical status.
3. Heart UK's activities may focus on individual level activities, individualising responsibility for CHD and detracting from the wider socio-economic conditions. This, however, cannot be attributed to an increasingly geneticised discourse about heart disease, but is a continuation of a more long-standing approach to health promotion and disease prevention.
4. HEART UK shows little interest in genetic technologies

Cox and Starzomski (2003: 162) argue that one of the reasons for the absence of geneticisation in the area of PKD is the 'lack of disease-specific support groups'. The findings presented in this chapter suggest that even where such groups exist, as in the case of FH and HEART UK, this is no guarantee of geneticisation. This highlights the value of studying patients' associations as sites that contribute to the construction of particular disease identities or cultures.

## CHAPTER 6: DEFINING AND EXPLAINING THE PROBLEM

### 6.1 INTRODUCTION

The following two chapters present the analysis of the interviews undertaken with lipid clinic patients who have FH. This chapter looks at interviewees' general accounts of the areas of FH and CHD. It focuses on how FH, hypercholesterolaemia and CHD are explained, and the underlying causal models implied by interviewees' accounts of these areas. In particular, it draws out the elements of these accounts where distinctions are and are not made between people with FH and others with raised cholesterol; and it draws attention to the degree to which genetics and genetic elements feature in these accounts of raised cholesterol and CHD. The following chapter considers the implications of this condition for the lives of these interviewees and their kin. Following Murphy (2000: 303) the interviews were seen as occasions in which the interviewees were engaged in 'constructing themselves, for themselves and for the interviewer, as normal, moral [and] responsible' in relation to their condition. The analysis, therefore, focuses on the *function* of what people say in the interviews.

The first two-thirds of the chapter discusses how interviewees explain FH and their accounts of hypercholesterolaemia and of CHD. The last part of the chapter focuses specifically on interviewees' constructions of the hereditary aspects of their condition. The chapter argues that the interviewees drew a firm boundary between their own *hereditary* raised cholesterol, which was only

controllable by medication, and other people's *lifestyle-related* high cholesterol. It is suggested that, within the prevailing culture of personal responsibility for maintaining health, and particularly for avoiding CHD and high cholesterol, that this boundary allowed interviewees to establish their own lack of culpability for having raised cholesterol. Nevertheless, interviewees' talk about their condition was characterised by a great deal of reference to their own lifestyle, which was sometimes in direct contradiction with their characterisation of their raised cholesterol as being largely unaffected by lifestyle. It is suggested that the ethos of personal responsibility concerning cholesterol and CHD is so strong in Western culture that interviewees do not necessarily relinquish it. Instead, they tended to oscillate between these different discourses. Lifestyle talk contributed to a wider theme, that FH could be controlled and CHD avoided by taking care of oneself. Although genetics was seen as a contributing factor, interviewees' accounts of their own and other people's CHD did not foreground the genetic elements, but conformed to established lay models of CHD encapsulated by the idea of the coronary candidate (Davison et al., 1989, 1991, 1992). The second part of the chapter suggests that FH was not associated with a strong disease identity and that interviewees may understand their condition as being a general category of hereditary hypercholesterolaemia, or high cholesterol that runs in families, rather than a specific genetic condition with quantifiable and predictable transmission patterns. The implications of such an understanding are considered in terms of the construction of risks and responsibilities.

## 6.2 NAMING THE PROBLEM

Interviewees had a number of different ways of referring to their condition. Some talked of familial hypercholesterolaemia or FH, and some expressed the meaning of FH in more manageable language, for example saying familial high cholesterol, family cholesterol or hereditary cholesterol. Others indicated that they recognised the diagnosis, at the same time as demonstrating the complexity of the name, for example: ‘familial whatever’, ‘high cholesterodaemia blah, blah blah, the inherited type’ or ‘hyper fam, no i can’t get my tongue round it at all’. Occasionally the idea of a specific diagnosis was absent, or the diagnosis was unmemorable. This is demonstrated in the following extract with a patient and her daughter-in-law (denoted as DIL):

KW: Right, and has anybody ever given it, sort of official name the high cholesterol problem?

ID87: Oh no, no

DIL: There is a name

ID87: ay?

DIL: There is a name

ID87: Yeah there is a name, um I don’t know what it’s called

DIL a long name

KW: a long name?

DIL: yes

ID87: er whatsit’ll tell you um, the lady I’ve just been to see now.

These data suggest that the specific diagnosis of familial hypercholesterolaemia is not necessarily particularly meaningful or memorable. This is not to say that the familial aspect of the high cholesterol was not a theme, but rather that FH, as a particular diagnostic category, does not have a high profile. This point is important in as much as it explains the relatively few references in the data presented to FH, and the more frequent talk of a cholesterol problem or condition. It also contributes to a suggestion

made later in this chapter concerning the lack of a specific disease identity for FH.

### 6.3 EXPLAINING FH

Every interviewee recognised an hereditary or familial aspect to their condition at some point during the interview, although the amount of focus and degree of significance placed on this element varied greatly. Interviewees' explicit accounts of FH, or their condition as they saw it, can be divided into three main elements:

1. **genetic/hereditary:** e.g. 'something that's passed on in the family', 'an inherited problem', 'a faulty gene'.
2. **too much fats/cholesterol in the blood:** e.g. 'furring up of the arteries', 'fats in the arteries that builds up', 'fats in the blood stream'.
3. **a problem of processing cholesterol:** e.g. 'body produces too much cholesterol', 'a missing enzyme to break down fats', 'liver doesn't get rid of cholesterol properly', 'cells don't excrete fats as they should'.

Interviewees often drew on a combination of these elements in their accounts of the condition. In some cases there was a very strong focus on the genetic elements. This is exemplified in the following extract, in which the interviewee draws on two elements to explain his condition, heredity and cholesterol processing, although the main emphasis is on the hereditary aspects:

KW: If you had to explain FH to somebody who had never heard of it before, what might you say?

ID77: I'd just say it's a genetic defect. I've inherited from me father. Basically your liver doesn't scrub out the cholesterol as it should do and

you've got a fifty per cent chance of inheriting it. I've inherited it and one in five hundred people suffer from it. And that's it, basically, that's all I'd say.

In other cases the genetic element was mentioned, but was less prominent.

Significantly, the genetic element could be entirely absent in accounts of the condition. The following explanation, for example, refers only to a problem of processing cholesterol that results in too much cholesterol in the blood:

KW: Let me ask you now, if you had to explain your problem to somebody who didn't know anything about it, what would you say to them?

ID87: Well I usually say, what it is, it's um your body makes too much cholesterol and when you've got too much cholesterol, it clogs the arteries and if you smoke and you've got high cholesterol you could have your legs off, could turn to gangrene, um heart attack, could be fatal heart attack, could have a stroke.

In some cases the account of FH given depended on who the interviewee imagined they were talking with. For example, ID24 constructs FH as a liver condition, without mention of genetics, in his account of how he might explain the condition to someone who had never heard of it. His account of what he told his children about the condition was similar in his focus on a liver problem, yet on this occasion the hereditary aspect was mentioned. The two accounts are provided below for comparison:

KW: If you have to explain FH to somebody who's never heard of it, what would you say?

ID24: I would say I've got a condition that's affecting my liver and, I mean we produce natural cholesterol fats in our bodies and we have them in our bodies through our diet. I would say that my liver should be able to remove them naturally and my liver does it probably at a much lower efficiency than everybody else's. So I would say it's probably a condition of the liver, that it's not removing cholesterol or breaking down cholesterol as it should.

KW: So what did you say to your kids?

ID24: Well I told them what the problem was as far as I was concerned, I just simply said, well I've got this genetic problem connected with my liver really, my liver's not removing cholesterol as it should do and

there's a chance that you've got it, I said, and the earlier you get diagnosed for it the more chance you have of sorting it out.

One possible interpretation is that the genetic element of FH is seen as stigmatising, and therefore this element might be left out of accounts of FH for a general audience. However, stigma was not a major theme in these interviews (this will be discussed in more detail in later sections). An alternative interpretation is that the hereditary aspect of FH is of variable relevance and in some situations is less important. In any case, these data demonstrate that the genetic element of FH is not always at the forefront of accounts of the condition. It can also be constructed predominantly as a liver or metabolic problem or as problem of high cholesterol.

#### **6.4 EXPLAINING HIGH CHOLESTEROL**

Interviewees talked about different types of cholesterol. There was 'my type of cholesterol' or 'the inherited type of cholesterol', and there was 'ordinary cholesterol'. These types of cholesterol can be related to the causes of high cholesterol, which were, in the main, divided into two categories: either genetic/hereditary, meaning FH or *my condition*; or dietary, meaning *everyone else*. FH or *my condition* can be typified as being a condition that **cannot be controlled by diet and requires medication**, in contrast to other high cholesterol, which may be controlled entirely by diet or other lifestyle modifications. This seems to be the same distinction between hereditary hypercholesterolaemia and lifestyle-induced hypercholesterolaemia that is evident in HEART UK's literature and in the biomedical literature, discussed in the previous chapters.

The construction of these two contrasting classes of high cholesterol, here framed as FH or my condition versus dietary or everyone else's, reflects a very strong discourse of culpability and personal responsibility concerning raised cholesterol and CHD in both lay and professional discourses (Davison et al., 1989, 1991, 1992; Lupton, 1995; Lupton & Chapman, 1995; Petersen & Lupton, 1996; Sachs, 1996). This distinction between my high cholesterol and dietary high cholesterol allows the interviewees to distance themselves from potential reproach for unhealthy living. This was also evident in the recurrent use of a phrase concerning FH that 'there is nothing you can do about it'. This was mostly used in the sense that one cannot avoid having FH, but was also used to mean that if one has raised cholesterol due to FH, there is not much one can do through altering behavioural regimes, to lower this cholesterol. These points are illustrated in the following extract. Here the interviewee was talking about how she explains her heart disease and cholesterol problem to her friends and workmates:

ID18: I just say all I know that it's to do with the fats. There's good fats, there's bad fats or whatever and some people, cholesterol is in your body anyway, I mean it's one of them things and it's ordinary cholesterol, you can get it down by diet, but for some unknown reason this type of cholesterol I've been told is hereditary and there's not really a lot I can do about it and it has to be done by tablets.

The interviewee clearly distinguishes between *ordinary* cholesterol, which one can *get down by diet*, and hereditary cholesterol, which *there's not really a lot one can do about*, so it has to be managed through medication. In drawing a distinction between ordinary cholesterol and her own type of cholesterol the interviewee establishes that, in her case, she is not responsible or to blame for her raised cholesterol.



The censure of ‘ordinary’ hypercholesterolaemia is implicit in the interviewees’ accounts of the causes of their high cholesterol. This is illustrated particularly well in the following extract, which was a response to a question about why the interviewee thought she had raised cholesterol:

ID104: It’s genetic basically, I hope. I know a little bit you can help it along by eating the right things and regular exercise, but apart from that it’s genetic.

The use of the phrase *I hope* in this context implies that it is better that raised cholesterol is due to genetic causes rather than due to behaviours that are ostensibly within one’s own control, like *eating the right things* and *exercise*. The role attributed to diet and other lifestyle factors in FH or *my condition* varied, but, as the extract above suggests, was mostly constructed as minor or adjunctive and certainly not sufficient to treat the condition. The idea that FH or the interviewee’s condition cannot be controlled by diet and requires medication was sometimes substantiated by reference to their own experiences of the management of their high cholesterol. In the following extract, when asked to explain her cholesterol problem, the interviewee’s immediate response was to provide an account of her initial attempts to reduce her cholesterol levels through dietary changes:

KW: Um what sort of ideas do you have about why you’ve got the cholesterol problem?

ID97: First of all, definitely ‘cos of the genetic factor because with diet it went down from 9.2 to 7.9 and that was me pulling out all stops with diet. You know really sort of, I had a food diary. I wrote everything down that I ate and I was good. So I knew I couldn’t get it lower than 7.9 and live happily, with just diet so I knew it must be a faulty gene.

This account allows the interviewee to demonstrate the effort and diligence with which she attempted to control her cholesterol level through dietary

changes, firmly establishing that, in her case, the limited reduction was not through any lack of trying. It is notable that the interviewee's conviction that cholesterol is controllable by diet is not shaken by her own reported experience of diet making only a marginal difference to her own cholesterol levels. This serves, rather, to reinforce the genetic causes in her own case. There is little space for other possible causal elements or for the possibility that dietary modifications may be of variable efficacy for other people.

There were occasional exceptions to this heredity versus lifestyle model of hypercholesterolaemia. For example, four of the interviewees attributed a larger role to dietary causes in their own raised cholesterol. It was notable that these were all somewhat older interviewees and that, in all cases, discussion concerned past rather than present behaviour, which allowed the interviewees to distance themselves from their earlier 'errant' dietary patterns. The following excerpt provides the most explicit example of these:

KW: So um what sort of ideas do you have about why you've got the cholesterol problem and why you've had the heart attack now?

ID102: I think it's eating fatty things. You see before I got married I used eat a lot of butter you know, used to cook in lard didn't they. They didn't have these cooking oils like they have then, and I think it's a lot to do with that really. Same with bacon. You'd eat all a round of bacon, you know so, I think it's not watching me diet years ago in't it that.

At other points in these interviews, genetic causes were drawn on to explain the raised cholesterol. This illustrates a more general theme of these interviews; interviewees expressed multiple and sometimes contradictory discourses about FH. This theme will be discussed in more detail later.

There was very little discussion of the possibility that other factors might be involved in raised cholesterol. Two areas that were mentioned were age and gender (these same areas were also mentioned by HEART UK interviewees, as discussed in the previous chapter). The idea that raised cholesterol is something that people develop as they get older emerged in a small number of interviews. Firstly this happened by comparison, which can be typified as *you don't expect someone my age to have raised cholesterol because it's an old person's condition*, and secondly by reference to interviewees' own parents developing raised cholesterol later in life:

ID79: my mother, she's 85, she's incredibly fit and amazing, but she's got a little bit of cholesterol, but I think it's just age possibly.

The relationship, or perhaps dissonance between the diet/lifestyle discourses and this age discourse was not discussed or problematised in any of the interviews.

The second area concerns gender differences. This is not so much that men and women have different levels of cholesterol, rather that raised cholesterol has a differential impact. It was suggested that men with FH are likely to get heart disease earlier than women or that women are less susceptible to the disease. The mechanism for this was related to women's hormones, and the menopause was mentioned as an important life point after which cholesterol management might become more of a concern for women.

So far, this section has argued that interviewees largely divided the causes of hypercholesterolaemia into two categories, hereditary/my type of cholesterol

and ordinary/lifestyle-induced cholesterol. It is notable that the interviewees' own experiences of an hereditary basis to hypercholesterolaemia do not appear to lead them to question the link between diet and raised cholesterol for other people. Indeed, the distinction between dietary and hereditary hypercholesterolemia perhaps becomes more entrenched in interviewees' accounts, as part of the process by which they establish their own lack of culpability. This contrasts with Lupton & Chapman's (1995) study of lay constructions of hypercholesterolemia, which drew on a sample from a general population. Although this sample strongly linked hypercholesterolaemia to diet and body size, Lupton & Chapman (1995) reported considerable uncertainty and complexity in their accounts. The difference between these studies is perhaps that Lupton & Chapman's (1995) interviewees were talking of hypercholesterolaemia in general, whereas in this study the interviewees were obliged to account for their own hypercholesterolaemia, a situation which required them to demonstrate their sound moral standing.

Since high cholesterol may be thought of as stigmatising<sup>13</sup> in the sense that it is associated with lack of self-discipline and failure to behave appropriately, one might expect this to be a theme in interviewees' discussions of their own experiences. Yet, this was almost absent from their accounts of anticipating people's responses to their high cholesterol. According to interviewees, in the main they were happy to tell people about their raised cholesterol, with little indication that they expected to be judged for it. My question about whether the interviewee had ever been in a situation where they wanted to avoid talking

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<sup>13</sup> The issue of whether the genetic aspect of FH is seen as stigmatising, as opposed to the raised cholesterol aspect, will be discussed in the next chapter.

about their condition mostly elicited a short answer such as ‘no, not at all’.

This is a typical response:

KW: And are there any situations where you’d avoid talking about having high cholesterol?

ID29: No if somebody asks me a question I’ll tell’em all I know, like about diets and foods, best I can you know what to do.

KW: So there’s nothing embarrassing about having high cholesterol?

ID29: No I wouldn’t think so, why should there be?

However, lack of stigma concerning their condition was also related specifically to the interviewee’s lack of agency for their raised cholesterol. In the following extract the condition is framed as not embarrassing because there was *nothing I could do about it*. This implies that raised cholesterol may be stigmatising where it is seen as due to a person’s individual volition:

KW: Alright, are there any situations where you would prefer not to talk about it or you would prefer to avoid talking about it.

ID79: no, it doesn’t embarrass me or anything like that as I say because I took it straight away it was something I could do nothing about, that I was born with it and that’s it.

Nevertheless, only one interviewee explicitly acknowledged that one might be judged for having raised cholesterol. This young man reported that he was now more careful about telling people than in the early days because he was wary that everyone assumed he ‘doused everything in lard kind of thing’ or that ‘my mum didn’t feed me healthy food’.

While accounts of FH and raised cholesterol are based on a model that involves a high degree of personal responsibility, interviewees rarely blamed other people with raised cholesterol outright for this condition, although, occasionally, explicit blaming language came to the fore. For example, one woman distinguished her family high cholesterol from something ‘you do to

yourself through bad-eating' and another man contrasted his own heart disease caused by FH with a colleague's, which he attributed to 'fatty dinners and smoking and sitting in the car all day'.

Overall, there was clearly a discourse within these interviews of responsibility and culpability associated with raised cholesterol. This was evident, for example, in the construction of two distinct classes of raised cholesterol and the use of the phrase 'there's nothing you can do about it'. This distinction emphasised the idea that 'ordinary' cholesterol can be controlled through lifestyle measures, whereas FH or my condition cannot. The ambiguity and uncertainty reported in other studies such as Davison et al., (1989, 1991, 1992) and Lupton & Chapman (1995) are absent here. This may be because the distinction is necessary to allow interviewees to defend themselves against imagined accusations of inappropriate behaviour. Nevertheless the language of culpability seemed to dissipate when interviewees talked about accounting for their condition to other people. Given the strength of discourses of responsibility, it is surprising that this area of accounting for one's high cholesterol to others, of avoiding blame or being required to refute it, gained such little attention during the interviews.

## **6.5 LIFESTYLE TALK**

Despite the distinction interviewees made between their own hereditary hypercholesterolaemia and other people's lifestyle-induced hypercholesterolaemia, all but two interviewees discussed or made mention of their own diet or dietary and lifestyle modifications they had made, and a

number of interviewees spoke at length about these. This section will suggest that interviewees tended to emphasize lifestyle issues in their talk about their experiences of having FH and that this can be situated in the framing of CHD and hypercholesterolaemia as moral issues, strongly linked to personal responsibility and culpability concerning 'lifestyle'. The section proposes that the power of moral discourses about diet and lifestyle in Western society are such that, in spite of the availability of genetic explanations in the case of FH, and the discourse of the limited impact of lifestyle compared with medication, interviewees still found it necessary to demonstrate their moral probity vis-à-vis lifestyle factors.

The strength and significance of lifestyle talk for constructions of FH is illustrated particularly well through interviewees' discussions about the impact of the condition. Prior to the interviews, I anticipated that this might be framed in any number of ways, for example, in terms of the family, life plans, illness experienced or possible future disease, future uncertainty and so on. Yet, in a number of cases, the impact of FH was immediately related to diet and lifestyle, either in terms of changes made, or the lack of necessity for making changes because the interviewee already had an appropriate lifestyle:

KW: So what sort of impact would you say kind of getting this diagnosis has had on your life?

ID90: Well dietary, my diet's had to change, I mean it's partly been a good thing, but once, I think once I was used to changing the sort of way that I ate to be honest, you sort of go up a level, and then it just becomes normal after a while. So not having butter, not having full-fat milk and not having eggs, and not having a boiled egg for breakfast, which I did everyday, after a while it just, I don't even notice.

KW: um okay what sort of impact would you say having this familial hypercholesterolaemia has on your life?

ID100: I had quite a healthy lifestyle and I'd been brought up with a mother who years ago when you had high cholesterol you went on a low fat diet, so I've been brought up on it. We never had butter, do you know what I mean, so it really didn't change the way I lived like that, because that's how I was brought up and that's how I lived.

The connection between lifestyle and raised cholesterol was so strong that some interviewees even said that having to make dietary changes had been the worst aspect of their condition, and this included people who already had

CHD:

KW: What would you say are the worst aspects of having it?

ID24: Well the worst aspects I suppose are, it's things like, it's, I love cheese for example, I really do like cheese and I have to limit what I take. You know I do, when I eat cheese, a small bit of cheese, I enjoy it, but I could have twice as much.

While interviewees' explicit accounts of the causes of their raised cholesterol attributed medication priority over lifestyle, their talk about their own behaviours and responses to the diagnosis could give equal or even greater focus to diet than to medication. In the following extract medication is mentioned, yet diet and lifestyle seem to be constructed as the critical factors that could mean the difference between health and ill health:

KW: What sort of impact would you say that it's had on your life?

ID21: Well I wouldn't say that it's had a big impact. The only impact it's had is like diet, have to watch what I eat you know, fat-wise and stick to low fat diet, just having to take medication's the only impact I can see, yeah.

KW: And do you think of it as a serious condition?

ID21: I suppose it can be if you didn't watch yourself. You know, I mean if I was to go mad and not follow, like the diet sheet that I've been given to watch what I eat and all that, I suppose I could be a lot less healthier than I am, because I really look after myself, knowing I've got this condition, and knowing it could be serious if I didn't take care of myself and what I eat and drink and all that.

This extract suggests that the interviewee is engaged in showing herself to be a responsible person, who *takes care of herself*. This is entirely congruent with



an ethos of individual responsibility for health and illness. The interviewee's sound moral status is demonstrated through reference to adhering to an appropriate diet. Medication commands relatively little attention, yet responsibility for self-care could equally have been framed in terms of responsibility for taking medication appropriately. This suggests that lifestyle remains an over-riding moral concern.

On occasions interviewees' discourses appeared to be plainly contradictory regarding the importance of lifestyle factors compared with medication. For example, when I asked ID80 how he might explain his condition to other people, he provided the type of account discussed in the previous section, focussing on the need for medication and the relatively negligible impact of diet and exercise:

ID80: The only understanding I've got is your body just produces all this cholesterol and it doesn't matter really 'ow much you diet or 'ow much y' exercise, your body'll just carry on producing it. So even though diet and exercise can 'elp a little bit, it's just basically like, it's just barely touching it. You could never drop it down from, say my 14.3, I could never ever get that down no matter 'ow much dieting or exercise I do, you can never ever gerrit down, 'cos your body just carries on producing it. So it can only be reduced through medication.

Nevertheless, this interviewee devoted a great amount of time in the rest of the interview to talking about his lifestyle, including a lengthy account of the dietary changes he had made as a result of diagnosis, a discussion of his changed drinking habits and the exercise he takes. Here is an example of this recurrent lifestyle talk in this interview:

KW: And um when you think about your own future health now, how d'you see it?

ID80: Er I need to get fitter and loose some weight (laughs), so I've joined a gym so I just try and keep me fitness levels [discussion of activities at gym] but I just wanna try and lose weight. If I can lose a

stone I'd be 'appy [...] I'm determined I want to get fitter and if I can lose about a stone in weight er it's not gonna be easy but I'll do it. I look after meself better now.

KW: And do you think by doing that, that'll help you to not have more heart problems?

ID80: Yeah definitely and also, me diet's a lot better as well now than it used to be, you know, 'cos a lot of the times it used to be, er well not a lot of the time, but on a fairly regular basis used to be easy option for your tea, you know, lets go to the chippy, or whatever [...] So we eat we eat better food, more healthy food than we used to do, so continue doing that.

Here again, questions about future health are used as an occasion on which to demonstrate that the interviewee is taking care of himself by adhering to particular fitness and dietary regimes. Again lifestyle is constructed as the critical factor in maintaining health, even though this man asserts elsewhere in the interview that medication is the critical factor.

These data suggest that interviewees had multiple and sometimes contradictory ways of framing FH, which emerged at different points in the interview, and that these were tied up with interviewees being able to present themselves as morally sound in relation to their condition. This involved, on the one hand, refuting responsibility for having hypercholesterolaemia through reference to an hereditary basis which decoupled raised cholesterol from lifestyle in their own case, but, on the other hand, also taking responsibility for looking after themselves, which was framed more in terms of adherence to appropriate lifestyle rather than to taking medication. Issues of lifestyle and diet retain a high position for FH patients, despite claims that these factors had relatively little impact. This reflects the strong, seemingly incontrovertible link between diet/behaviour and personal responsibility for raised cholesterol in health discourses in contemporary Western culture. This discourse is so dominant

that interviewees do not necessarily relinquish it, even when faced with knowledge of a genetic aetiology that might allow them to do so. Instead they tend to hold multiple discourses, weaving back and forth between them.

## **6.6 TAKING CARE OF ONESELF**

The previous section demonstrated the strength of lifestyle discourses in constructions of FH. These contribute to a wider discourse concerned with taking care of oneself. A strong theme in interviewees' talk about FH was the controllability of this condition and the avoidance of disease through taking care of oneself by adopting an appropriate lifestyle, taking medication and attending appointments with health care practitioners:

ID16: It could be a serious condition if I did nothing about it or if I disrespected everything that people tried to do to make my life last longer and be better. But provided I stick to the guidelines then I hope I, respecting everything that people are doing for me, I'm able to extend my life to be healthy.

One could argue that these discourses of avoiding CHD through taking care of oneself conform to Novas & Rose's (2000) notion of genetic prudence i.e. the responsibility to manage oneself in the light of genetic risk information. However, in this case it is difficult to tease apart the degree to which this imperative to take care of oneself is related specifically to genetic risks or to more general personal responsibilities that might apply to any person deemed to be at any kind of risk of any condition. The idea of genetic prudence is based on the provision of genetic risk information in the absence of physiological measures. In the case of FH, however, risk of future CHD is based on measuring cholesterol levels. In this respect it would be illuminating to compare these constructions with how people with 'ordinary' or 'polygenic'

hypercholesterolaemia, or perhaps with high blood pressure, construct their condition and the attendant risks and responsibilities.

Taking care of oneself was constructed as an area of *responsibility within reason*. This was concerned with avoiding disease whilst managing the practicalities of every day life or maintaining a reasonable quality of life. This idea was sometimes expressed in a confessional language such as ‘pinching a biscuit’, or not being ‘a monk’, ‘a virgin’ or ‘perfect’. It was important not to be too ‘anal about things’ or to ‘take things too seriously’. In the discourse of ‘responsibility within reason’ a balance is required between avoiding disease though taking care of oneself properly, and maintaining a reasonably normal life. It is possible to take FH both too seriously and not seriously enough. This was demonstrated neatly by one woman’s discussions about herself and her siblings. On the one hand, her sister died at the age of fifty of a ‘massive heart attack’, because she would not take it seriously enough; she was ‘overweight and everything that she shouldn’t do, she still continued to do’. There was also a question mark about whether she actually took her medication:

ID33: Whether she took it or not, she was a bit dizzy (laughs), lovely, but wouldn’t take anything seriously, would she?

On the other hand, her brother ‘takes his medication too seriously’ and is ‘a bit obsessive’ about his cholesterol level. The interviewee’s own view was that you have to get ‘a happy medium’:

ID33: alright you’ve got a problem, you can’t override your life. You know you deal with what you have to deal with it and then you forget about it.

This echoes Backett's (1992) work on lay health moralities, cited in Lupton (1995), which found that 'being obsessive about one's health [was] just as much a taboo as being overly cavalier' (Lupton, 1995: 140), and others who have suggested that lay people are cognisant of public health messages, but weigh these up in terms of the practicalities of everyday life (Lambert & Rose, 1996; Williams et al., 1995).

There was only one exception to this discourse of controllability and avoidance of disease, where an interviewee expressed explicitly fatalistic ideas about her condition:

KW: the idea of getting heart disease, is it something that worries you?  
ID04: It does in one way, I just kinda think, oh well it runs in the family, it's gonna 'appen to me anyway, so if it 'appens, it 'appens kind o' thing. But it just, if I really think about it, it does scare me a little bit, but I just kinda think, oh well I'm one of them that are gonna go early, if the rest of the family 'ave, so.

This section has suggested that FH was constructed as controllable and CHD modifiable by *taking care of oneself* and that actual cases of CHD were explained within this discourse. The following section discusses interviewees' accounts of CHD more widely.

## **6.7 EXPLAINING CHD**

Interviewees' accounts of CHD confirm a role for genetics in the case of FH. However, the degree to which CHD was attributed to genetics for people with FH varied between interviews. In some cases this was offered as the main cause. In the following example a woman who does not have CHD accounts for her sister's CHD. We have already established that her sister has FH:

KW: So why d'you think she's got the heart problems?

ID19: She must have more of the genetic makeup, I may have more of my mother's genes than she has. Or I could have more my dad's father's family's genes. She's obviously got more of the maternal makeup, my grandmother's family's make up in her<sup>14</sup>.

In other interviews, CHD was attributed to a combination of factors, which were all recognisable aspects of general lay constructions of CHD, encapsulated by the idea of the coronary candidate (Davison et al., 1989, 1991, 1992). This included factors such as temperament, stress, appearance and behaviour. In the following example a man discussed his father's CHD:

(Para 15)

ID77: My father died when he was forty-seven. He wasn't particularly overweight, even though he was overweight, but not particularly obese. He wasn't particularly fit, but even so, forty-seven was too young.

(Para 43)

ID77: Got married when he was thirty-two, was slim up to that point. I don't know what kind of exercise he did at that point. But he's obviously, by the time he's forty-seven he is overweight, did he say fifteen, sixteen stone. I think he was six foot, five foot eleven I think he was, overweight, but not excessively, drank too much beer. He had a good life, enjoyed himself completely, no doubt about that, had a kind of fiery temperament, did have a bad temper.

In this interview, although the father's early death requires some additional explanation, the account of his CHD still draws on ideas about weight, fitness, alcohol consumption and temperament. Even where interviewees explained their own CHD, they might still draw on a wide set of factors. One woman, for example, suggested that her own CHD was due to: raised cholesterol; the stress of bringing up her children more or less on her own, working and 'rushing about'; and being 'very highly strung'. Interviewees sometimes included admissions of inappropriate lifestyle in their accounts of their own CHD. In

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<sup>14</sup> This excerpt indicates another theme concerning how inheritance patterns for FH were constructed, which will be discussed later in this chapter.

the following example, although the interviewee makes clear that he was aware that heart problems ran in his family, this is not his main focus:

(Para 42)

ID88: Prior to that I did live a good life [...] at my heart attack point I was maybe two stone overweight. So you know, I knew the family history, that I was sat on a time bomb, but at the end of the day, you know I was smoking as well, so as I said to you I had all the qualification, I was a highly qualified candidate.

(Paras 60-62)

KW: If you had to say why you had the heart problems and why you think you got it in '96, what would you say to that?

ID88: Er not taking head of things and not stopping smoking, eating the wrong things, not taking much exercise, um and generally being blasé towards life.

The role attributed to genetics varied not only between interviews, but also within them. This further illustrates the observation that interviewees wove between multiple discourses. One woman, for example, accounted for her CHD entirely in terms of her raised cholesterol and, by implication, genetics:

KW: D'you think there was anything specific that brought on the angina at the time that it came on?

ID33: No, I think it was the cholesterol deposits that would've, what ever would've gone on in my life, wouldn't it? It would've come along.

Elsewhere in the interview, however, she talks of lifestyle and stress, suggesting a more multifaceted aetiological model:

ID33: We read up on both our conditions and er got to grips with it and tried to cut, you know, a lot of stress out of our lives, didn't we? Changed our diets completely and, well we both feel that if we hadn't've done, we'd both be in um, you know, a pretty poorer state than we are now.

These data reinforce the construction of CHD as avoidable through taking care of oneself. They also reinforce the suggestion that interviewees were obliged to oscillate between distancing themselves from responsibility for having high cholesterol, and showing that they, nevertheless, maintain a healthy lifestyle.

So far this section has discussed interviewees' accounts of CHD for people where a diagnosis of FH was established or suspected. In the following example, an interviewee provided an account of early CHD that was not related to FH. This woman understands that her FH comes through her mother's side. Coincidentally, according to her account, her father had heart disease, which started with a heart attack at the age of thirty-six. She explains this as follows:

ID60: The fact was my dad's mum left him when he was about nine and that's when the dad [...] came back and started looking after my dad and he would have had fish and chips and really extremely bad nutrition, right from the word go. And then my dad went into a trade and became a brick layer. So it would be fish and chips everyday and it was only when my dad was first thirty-six, when he had his heart attack and my cholesterol was discovered, um that our diet changed [...] So I would say that, bad luck and whatever genetics that are in him contributed to it. But I would say a really bad diet contributed to my dad.

This account mainly draws on the ideas of the manual trade and poor diet, leaving some space for luck and genetics in general. Given this interviewee's own diagnosis of FH, I find it surprising that she does not draw more strongly on genetics or heredity to explain her father's early CHD. In an earlier section, it was similarly suggested that interviewees do not seem to draw on genetic factors in explaining raised cholesterol, other than in the case of FH, or 'my condition'.

In summary, interviewees' accounts of CHD in the case of FH are not necessarily fixed on the genetic elements. Furthermore, experience of FH does not appear to mean that interviewees adopt more genetic models in accounting for raised cholesterol or CHD in general. Indeed, the coronary candidate appears to be extremely tenacious. Davison et al. (1989) argued that in this



model, areas of non-control are seen as subsidiary to lifestyle. This does not appear to be radically changed through knowledge and experience of FH.

## **6.8 CONSTRUCTING THE HEREDITARY ASPECT OF FH**

This thesis set out to explore the extent to which FH is constructed as a genetic condition and one part of this must be to look at the ways in which or sense in which interviewees constructed the hereditary element of their condition. This section will focus on the ways in which people identified their condition as a family or hereditary condition and on their understandings of the transmission of the condition between generations.

### **Disease Identity**

The previous chapter concerning HEART UK suggested that there was not a strong FH identity and this is supported by the data from the interviews with lipid clinic patients. This contrasts with findings from other studies of genetic conditions, such as Richards (1996a) and Cox & McKellin (1999), which suggest that, particularly for dominantly inherited conditions, an awareness that the kinship may be 'prone' to the condition may become part of 'family culture'; it becomes 'this thing in our family' (Cox & McKellin, 1999: 629). In the case of FH, also a dominantly inherited condition, the minority (about a third) of the interviewees talked about a family narrative of a family history of CHD or raised cholesterol. One very clear example was provided by ID100, who explained that she came to be a patient at the clinic because she noticed that she had 'deposits' above her eyes and that she recognised them because

her mother, aunt and grandmother, who all have angina, also had them. She explained that she was not surprised by her own diagnosis of FH:

ID100: Because I knew my mother had it. It was a thing I'd grown up, d'you know what I mean, although there was no label to it.

In the remainder of the interviews either family history was simply not discussed or interviewees talked of piecing family health histories together after their own diagnosis was established. ID11, for example, said that she found out that she had raised cholesterol when she was thirty through having a 'lump' removed from her ankle, and was diagnosed with FH at the age of forty-four. She suggests that it was only at this point that she started to recognise the significance of her mother's own health history. Here she describes how she came to realise that her mother almost certainly had FH:

ID11: my mother wasn't one for talking a lot about things and she had big lumps on her hands which she wasn't bothered about because they never hurt her. She never actually said she had high cholesterol, she just took these tablets that they gave her at the hospital. It was only when I started taking them that I realised that they were the same ones that she'd been on, but because by then she'd passed away.

These data support the suggestion by Lambert & Rose (1996) that the hereditary aspect of FH often only becomes significant retrospectively, once the diagnosis of FH has been established.

The idea of a lack of FH identity or family narrative of FH is further supported by interviewees' accounts of how they came to be tested for cholesterol. These can be grouped into four categories, which are outlined in Table 6.1

**Table 6.1: Route to diagnosis of high cholesterol**

	<b>Number</b>	<b>Per cent*</b>
Death, CHD, raised cholesterol or FH in relative	12	40
Outward signs of cholesterol	9	30
Opportunistic testing	7	25
Own CHD	3	10

\* per cent rounded to nearest 5.

The largest group (40%) decided, or were advised, to take a cholesterol test as the result of the onset of CHD or death in a relative, or the diagnosis of raised cholesterol or FH in a family member. The second largest group (30%) were referred for cholesterol testing as a result of consulting about lumps that were bothering them, on the ankles, knees, knuckles and around the eyes. These lumps turned out to be cholesterol deposits, although to the interviewees this connection between lumps and raised cholesterol was unexpected. One quarter of the interviewees reported that they found out they had raised cholesterol through opportunistic testing, for example through a workplace testing scheme or primary care ‘MOT’. A small number found out about their raised cholesterol only as a result of being treated for CHD. Overall, the diagnosis of raised cholesterol often came about by chance and was unanticipated for a large proportion of these interviewees. As exemplified by ID11, prior to diagnosis of raised cholesterol, and even after, interviewees did not necessarily have a strong sense of a family history of CHD.

### **Understandings of the hereditary transmission of FH**

This section focuses specifically on how interviewees understood the hereditary aspects of their condition. This did not constitute a particularly large part of patients’ talk and the interview schedule did not contain any specific questions in this area. While it was a relatively minor aspect of the

interviews, it is nevertheless important to draw out this data in order to get a sense of the ways in which interviewees constructed their condition as hereditary. It has already been suggested that, for the interviewees, FH does not have a high profile as a particular diagnostic category. Even where interviewees use the terms familial hypercholesterolaemia or FH, it is not self-evident what they understand by this. This section will consider in more detail how the hereditary aspects of the condition are discussed, whether and in what sense this is understood as a particular single gene disorder and the interviewees' understandings of the transmission patterns of the condition.

The aim here is not to 'test' interviewees' models of heredity against the 'correct' biomedical models. Indeed, as Emslie et al. (2003) comment, and chapter four of this thesis illustrates, biomedical knowledge about inheritance is not fixed, but can be contested. The aim is rather to consider the way genetic risks and responsibilities may be configured in relation to interviewees' understandings. The notion of genetic responsibility involves not only an ethos of responsibility for taking care of oneself, but also involves people constructing linkages between themselves and other people such as potential or actual partners, offspring and wider kinship. How these linkages are constructed, therefore has implications for the degree or sense in which genetic responsibility is assumed. The section suggests that in contrast with biomedical accounts, and possibly the expectations of analysts such as Novas & Rose (2000), a large proportion of interviewees may see the transmission of their condition as somewhat sporadic or unpredictable.

Richards (1996a, b, 1997) has made a number of observations about lay understandings of heredity, drawing together several central recurring concepts and these have been more recently reinforced by Emslie et al. (2003). These observations are based on studies of general populations and clinic populations for genetic counselling services, particularly relating to Huntington's disease and hereditary breast/ovarian cancer. The present study, therefore, offers an opportunity to consider these ideas in a new type of population. Important lay concepts about heredity include the idea that a condition can 'skip a generation', that characteristics are linked together, so that children who inherit a condition are likely to resemble or 'take after' their affected parent either physically or in character, and that certain characteristics are gendered, passing from mother to daughter or father to son. Richards (1997) and Emslie et al. (2003) observe that lay people often use genetic terminology such as gene and DNA, but that this is not linked to particular technical understandings of these terms. Rather, this language stands for the whole area of heredity:

'in this context, 'gene' seems to be a term for the general concept of the biological transmission of characteristics between generations'  
(Richards, 1997:190)

Many of these observations about lay people's talk about heredity are reproduced in the present study. Interviewees had a number of ways of referring to the hereditary aspect of their condition, ranging from very general notions of heredity, such as 'a condition that runs in families' or 'a hereditary thing' to more specific references to a gene, such as 'a genetic defect in your body' or 'a defective gene'. About half of the interviewees talked of *a gene*, in the singular, and in perhaps seven or eight of these cases, the discussions included elements of a Mendelian account of FH, such as (1) it is a single gene

defect (2) the prevalence of the condition is one in five hundred people (3) offspring have a fifty-fifty chance of inheriting the condition (4) there are two forms of the condition, heterozygous and homozygous, and the homozygous form is much more serious (although these specific terms were not necessarily used).

ID19, for example, indicated that this condition involves a particular gene when talking about her daughters coming for testing, saying that ‘two of them have the gene and one hasn’t’. Later on she demonstrated several elements consistent with a biomedical account of FH, particularly that this is an autosomal condition, in other words it affects both sexes, and that each child has a chance of inheriting the condition. Her talk implies a technical understanding of chance, as a random event for each child, through her suggestion that this is independent of how the child looks and of the status of any other children:

ID19: I’ve obviously inherited some genetic factor that runs in our family on my father’s mother’s side and it’s passed on to either sex, but you’ve a fifty-fifty chance of getting it [...] because two of mine have and one hasn’t, and yet they’re all exactly the same [...] They don’t look the same, I mean they’re very different to look at the children, but it’s just this gene that’s floating your way or not.

It is not clear exactly what this group understood the term ‘gene’ to mean and I did not ask this, and there was no mention of any specific genes, such as the LDL receptor gene. Nevertheless, this small group of interviewees demonstrated that they understood FH as a Mendelian condition on some level. They had some notion of a specific single gene condition, connected to some model of the processes and substances of hereditary transmission.

In other cases it was much less clear what an interviewee meant when they talked of a gene, genes, genetic factors or an inherited condition, although sometimes it was obvious that that they did not have Mendelian understandings of FH. ID78 stands out as extreme example. She explains her condition using the language of genetics, which at first glance implies some technical understanding of the condition. The following excerpt suggests three things (1) it provides a process that connects a 'rogue' gene in the liver to raised cholesterol (2) it makes a connection between genes and DNA (3) It establishes that ID78 is the only one of five siblings who has inherited the 'rogue' gene:

ID78: Well it's a rogue gene in the liver, they say it's like you're in a chocolate factory and it's pumping out coffee creams. When you've got too much cholesterol, it's just working overtime on the coffee creams are pumping out. 'Cos in about fifty years time they'll be able to do gene transplants. I've got some blood sent off for DNA in London [...] So what happened was out of the five children, I'm the one who inherited the rogue gene.

However, she went on to draw on a number of ideas about inheritance that are clearly incompatible with a Mendelian account. For example, although she repeated several times that none of her siblings had inherited the 'rogue gene' she reported that two of her siblings and several of their children had received treatment for raised cholesterol and her talk suggests that this is hereditary in some way. The implication in the following excerpt, for example, is that although the brother does not have the rogue gene, raised cholesterol has somehow been passed onto his daughter:

ID78: My son's been tested. He hasn't got any high cholesterol and me brother's children've been tested. He's got two boys, clear, and a girl, she took statin drugs. So it seems it's all gone onto the women.

My own immediate assumption, based on the account provided by the interviewee, was that it is very likely that this brother and one of her sisters had

FH and I find the interviewee's ideas both surprising and confusing. One interpretation is that she sees heredity and genes as different things, belonging to separate domains. This means that the idea of a rogue gene can stand alongside more everyday ideas about heredity without causing tensions. This idea will be discussed again. The interviewee's talk, in any case, illustrates that some interviewees talked of genes and genetics without this being connected to a Mendelian model of FH.

A number of lay notions about heredity emerged in relation to the transmission of FH. These ideas fit better with understandings of FH as a more general hereditary or familial condition than as a specific and knowable Mendelian condition. These included:

**Gendered transmission of FH:** The idea that FH might affect a particular gender was suggested in four interviews, implicating both men and women. In these cases the suggestion was related closely to recent family experience. For example, one woman with six siblings reported that it was the girls who had got the condition, commenting that:

ID21: it's something that's hereditary and passed onto, sounds like the female side of my family.

In this case, the link to gender is purely observational. In other cases the link was taken to be a recognised and predictable fact, for example, one man commented:

ID90: Well, I understand from [consultant] it tends to run down the male line.



In contrast to Lambert & Rose (1996), it is notable that this gendered construction of FH was applied to both men and women.

**Skipping a generation:** Richards (1997) notes that the idea that a disease may ‘skip a generation’ is very common and provides one way to explain the intermittent appearance of diseases within families, particularly in conditions with incomplete penetrance<sup>15</sup>. FH is generally understood to have high or near complete penetrance<sup>16</sup>. This means that it is very unlikely for the high cholesterol to skip a generation and this is reflected in the interviews. Nearly everyone identified a parent who had raised cholesterol, who had ‘passed on’ the condition, and no one suggested that FH had actually skipped a generation in their own case. Nevertheless, the possibility that FH might skip generations was present in four interviews. In the following example the interviewee has indicated that the condition comes from his father’s family, that his father and grandmother had raised cholesterol and that a number of his father’s siblings died of CHD. Yet his talk suggests a much looser lineage:

ID85: You may not be aware that your parents have had cholesterol. Maybe they didn’t, maybe someone further along the line had high cholesterol [...] I couldn’t explain it, other than the fact that it’s in your genes and it’s from somewhere of the genetic line.

It seems that ‘skipping a generation’ is such a central idea in everyday understandings about heredity that it filters through in the case of FH, even though it rarely ties in with personal experiences in this case. The fact that ID85 and others draw on the idea of ‘skipping a generation’ suggests a

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<sup>15</sup> Penetrance is the relationship between having the mutation and having the condition or disease. Penetrance is said to be complete or 100 per cent in cases where everyone who carries the mutation develops the condition.

<sup>16</sup> Although there has been some discussion of the relationship between genotype and phenotype in the biomedical literature, as discussed in chapter four.

construction of FH as a condition that belongs to the general category of hereditary conditions or conditions that run in the family rather than as a specific Mendelian condition.

**Mixed or blended inheritance:** Although most people identified a lineage on one 'side' of their family for the FH, in three cases interviewees mentioned both parents. This implies a model of raised cholesterol as a result of the mixing or blending of their parents' genes, traits or pronesses, rather than the passing on of a gene from one parent. In the following extract, the interviewee talked about inheriting a deformed gene, at the same time as discussing the heart disease of both her parents:

KW: What sort of ideas do you have about why you've got the cholesterol problem?

ID04: Because I've got a deformed gene that doubles the cholesterol. It's hereditary, that's all I know (laughs)

KW: okay, no that's a really good answer (laughs), was that a surprise to you when you were told about it or were there things in your family background that'd made you think you might have a

ID04: Well no, because of me family background. Me mum died, that was heart problems that me mum died, she was only fifty-one. Me dad died of heart attack. So I wasn't really surprised when you know they turned around and said, well, you've got this hereditary thing.

Later on in the interview I asked again about the family history and she talked again about both her parents. This suggests that she understands her own raised cholesterol to be the result of an additive effect of both her parents. The excerpt also supports Richards' observation that talk of genes can stand for the whole area of heredity.

**High risk estimates:** While several people proposed that the chance of getting FH was fifty per cent for each child, on two occasions interviewees seemed to

suggest much higher risk in their particular situation. In one case this was based on the high prevalence of the condition within the family. The interviewee suggested that his son was very likely to have inherited the condition, since everyone else in the family seems to have got the condition. In the second case, the suggestion is that the higher the parent's cholesterol level, the greater the risk of the child having raised cholesterol. Again this was substantiated through experience. In the excerpt, the interviewee is commenting on how she felt about taking her young daughter to be tested:

KW: But I think, I mean I suppose it might've been traumatic the idea of having to take her, to find out one way or the other?

ID11: Er no, well I suppose yes because there was always that fifty-fifty chance, but I think in my head I thought, well with me having such a high one to start with, she probably would, and she did. She had a high count.

The extract shows how Mendelian knowledge (the fifty-fifty chance) and other ideas, in this case perhaps a more blended model of heredity, coexist.

**'Taking after' and health trajectories:** Both Richards (1996a, b, 1997, 2003) and Davison (1996, 1997) have commented on the grouping or linking of characteristics in lay talk about heredity, suggesting that who someone is thought to look like or take after may be used as a basis for predicting future health trajectories or making risk assessments concerning specific genetic conditions. It was notable that there were only two examples of talk of 'taking after' in these interviews, both by people who had demonstrated a relatively biomedical understanding of FH. Taking the case of ID06, a man who works as a health professional, he observed that his young son 'is said to be more like me, I wonder if he might also have it'. There was no further talk about this in the interview and he gave no impression that he might treat his son and

daughter differently on this basis, or that he viewed his daughter to be less at risk.

**Mixed discourses:** The data presented support Richards' observation that many people talk about genes as a kind of short-hand for the whole area of heredity, rather than having technical understandings. This talk appears to sit alongside existing lay notions about heredity. Even those with relatively strong biomedical understandings of FH and genetics in general still also drew on other ideas about heredity, as in the case of ID06 discussed above. These data support the ideas proposed by both Richards (1997) and Emslie et al. (2003) that everyday ideas about inheritance and abstract biomedical knowledge about genetics may co-exist in separate domains.

In sum, this section has suggested that a number of recognised lay concepts about heredity were present in these interviews. While there were only a few references to any one of these, overall the data suggest that a large proportion of interviewees had some ideas that were at odds with a Mendelian model of FH. Perhaps half or more of the interviewees do not necessarily see their condition as one for which the mode of transmission is quantifiable and predictable. Rather, the condition belongs to a more general category of hereditary conditions or conditions that run in families that appear in a more sporadic or less predictable manner, or in the case of gendered constructions may be seen as only partially transmitted. This may have implications in terms of the way risks and responsibilities are constructed. These constructions of hereditary linkages must be coupled with other aspects of constructions of FH

which foreground individual actions and responsibilities. One might, therefore, speculate that for these interviewees, responsibilities based on genetic connections may assume less significance than might be anticipated through following analysts such as Novas & Rose (2000). This is borne out to some degree in the data concerning how interviewees construct responsibilities to others, which is presented in the next chapter.

## **6.9 CHAPTER SUMMARY AND DISCUSSION**

This chapter has argued that interviewees had multiple and sometimes contradictory strands in their constructions of their condition and that they tended to weave between these different strands. Interviewees explained FH drawing on three elements, heredity, a problem to do with high blood cholesterol, and a cholesterol processing problem. The genetic elements were not necessarily at the forefront of these accounts. However, they tended to frame the aetiology of raised cholesterol as a dichotomy, hereditary and only amenable to medication in my case, lifestyle-related for other people. At the same time, there was a strong degree of lifestyle talk in interviewees' discussion of their responses to their own cholesterol condition, which contributed to an overall ethos concerning FH that CHD is avoidable through taking care of oneself. The construction of the two distinct categories of raised cholesterol, and the fact that lifestyle talk remains prominent despite these constructions, reflects the seemingly immutable link between lifestyle and personal responsibility for hypercholesterolaemia in contemporary Western culture. In oscillating between different discourses at different points in the interview, interviewees managed to establish their sound moral status.

Interviewees presented multifactorial models of CHD that conformed to established lay constructions embodied by the notion of the coronary candidate (Davison et al., 1989, 1991, 1992). Genetics was attributed variable importance in these accounts, but was rarely seen as decisive in either their own or other people's CHD.

The chapter also suggested that FH is not associated with a strong disease identity. The terms familial hypercholesterolemia or FH did not have a high profile in interviewees' talk. Furthermore, the idea of a family history of CHD or cholesterol problems did not appear to have been an established part of family culture in the majority of cases. Interviewees' constructions of the specifically hereditary aspects of their condition suggest that many saw its transmission in less specific or predictable ways than suggested by a Mendelian model and it is argued that this may be significant concerning the way genetic responsibilities are constructed.

What can these findings contribute to the discussion of geneticisation? First genetics was not the dominant discourse in these interviewees' accounts of CHD. It is notable that even in this case, where people have specific knowledge concerning a genetic contribution to their own hypercholesterolemia, this did not lead them to foreground genetics in their accounts of their own CHD or other people's hypercholesterolaemia or CHD. Their constructions conformed to established lay models of CHD. Second, geneticisation suggests that society will become stratified along genetic lines. This was true in these interviews, in as much as interviewees distinguished

between their own hereditary hypercholesterolaemia and other people's lifestyle-induced hypercholesterolaemia. In this case, this enabled them to construct themselves as less, rather than more accountable for their condition. However, as already noted, in their general accounts of CHD, genetics was only one contributory factor, not a defining factor. Third, the great emphasis that interviewees placed on lifestyle and taking care of themselves could be interpreted as an expression of the individualisation of responsibility for preventing disease. However, as already argued, this is an established discourse concerning CHD and hypercholesterolaemia. Interviewees drew on constructions that already circulate in the lay population. This cannot be seen as a novel set of responsibilities imposed by an elite group of geneticists or other experts. These findings reinforce the argument that lay accounts as much as professional constructions tie health and illness to personal responsibility. In this sense, the findings support the critique of geneticisation that it underestimates the agency of lay people.

## CHAPTER 7: LIVING WITH FH

### 7.1 INTRODUCTION

This chapter is the second of the two chapters that draw upon the interviews with people with FH, undertaken at a lipid clinic. The chapter is concerned with what it means to have FH, how interviewees view their condition and their response in their everyday lives. The analysis focuses particularly on the ways in which FH is framed as a genetic condition through interviewees' talk about their actions in relation to FH. Rose and Novas (Novas & Rose, 2000; Rose, N., 2001; Rose & Novas, 2004) have argued that genetic knowledge creates obligations to consider decisions about the whole scope of life plans, which link individuals into a web of relations:

‘Choices about marriage, procreation, financial planning, inheritance, career and much more are made in a web of entanglements involving actual and potential kin, employers, partners and children’  
(Rose, N., 2001: 19).

One can argue that the degree to which people frame responsibilities in relation to genetic connections provides an indicator of the degree to which a condition is constructed as genetic. In other words, ‘genetic responsibility’ can be seen as an indicator of the genetic framing of a condition. If genetic or hereditary connections are at the forefront of people’s concerns about FH, then one would expect to see much talk in these interviews about people’s actions in relation to their kin, and other people to whom they are connected. The analysis is therefore concerned with interviewees’ talk about their actions in relation to others and the sense in which obligations are derived through genetic knowledge or connection.



The chapter first briefly reviews the notion of genetic responsibility and then presents data concerning the construction of responsibilities in relation to others. The main areas considered are reproductive decision making, taking care of one's offspring and talking with kin. The chapter argues that the construction of FH as highly treatable and manageable is used to distinguish it from other, more serious genetic conditions. This construction is central to interviewees' almost universal rejection of reproductive decision-making in the case of FH. However, there was a strong sense of responsibility for ensuring the appropriate testing, treatment and care of one's offspring. This can be seen as one aspect of genetic responsibility. There was a much less clear message about responsibilities to other kin, with only a small proportion of interviewees claiming responsibility for communicating information to their kin or for encouraging them to manage their cholesterol. Furthermore, interviewees also talked of informing other people in general about FH and cholesterol, and encouraging them to get tested or manage their cholesterol. These data suggest that responsibilities in relation to others are not solely or even predominantly derived through genetic risks and genetic connections.

## **7.2 GENETIC RESPONSIBILITY**

Genetic responsibility or prudence (Hallowell, 1999; Kenen, 1994; Novas & Rose, 2000; Rose & Novas, 2004) can be defined as the obligation to know and to manage the implications of one's own genome. It implies two areas of action. First, there is an obligation to become informed about one's genetic constitution and to undertake risk management to monitor and try to modulate one's own genetic risks. This is a continuation of discourses of personal

responsibilities to avoid illness through identifying health risks and adopting appropriate health-related behaviours (Petersen & Lupton, 1996). The previous chapter has shown that interviewees amply demonstrate these discourses, through constructing CHD as avoidable by *taking care of oneself*. Second, genetic responsibilities are extended towards other people, particularly family members. Hallowell (1999) and Polzer et al. (2002) have discussed the responsibility to disseminate information to relatives and encourage others to get tested, as well as to know and manage one's own risks for the sake of others. Novas and Rose's (Novas & Rose, 2000; Rose & Novas, 2004) notion of genetic prudence relates to a range of life plans, including forming partnerships, procreation, working life and financial arrangements. One can argue that recourse to reproductive decision making, in particular, has become integral to genetic ways of constructing conditions. In the UK, prenatal genetic testing and pre-implantation genetic diagnosis (PGD) are available for a range of Mendelian conditions such as Huntington's disease and cystic fibrosis, and consultation is underway concerning the extension of PGD for inherited susceptibility to cancers such as hereditary breast/ovarian cancer and an inherited form of susceptibility to bowel cancer (Human Fertilisation and Embryology Authority, 2005). This area could perhaps be thought of as emblematic of genetic ways of constructing disease.

### **7.3 REPRODUCTIVE DECISION MAKING**

In Chapter 5, it was noted that the idea of reproductive decision making in relation to FH was almost absent from HEART UK interviews and entirely absent from the biomedical literature analysed. This theme was similarly

unimportant to the FH patients interviewed at the lipid clinic. Discussions about reproductive decisions were mostly prompted by me and were almost universally framed by interviewees as a non-topic, i.e. they had been contemplated, but quickly dismissed, or were out with the bounds of what is thinkable about FH. This came through in discussions about interviewees' own children and in discussions about the potential of DNA-based technologies to lead to prenatal screening. The rejection of reproductive decision-making hinged on the construction of FH as treatable, manageable and not serious enough to warrant such a course of action. In the following extract, for example, FH was framed as something that is not necessary to consider when planning to have children, because it is a *manageable condition*:

KW: I was going to ask you when you were thinking of having your own kids whether it was something you thought about at all?

ID90: well we wanted to have children and the fact that you know I had this condition [...] and the fact that it was a manageable condition meant it was, to be honest, not really, it was a no-brainer. We wanted to have children, as long as we could.

The treatability and relative lack of impact of FH were underscored through comparisons with other diseases. The following extract again illustrates that reproductive decision making is outside the realm of considerations for FH, that this is because it is treatable and not that serious, and that this distinguishes it from other genetic diseases:

KW: do you think it would ever be a consideration for your children whether to have children or not, is it something that you'd have to think about?

ID15: No I don't think so. My son is not married but he's got a child from a relationship, he's now split up from the girl and I don't think it's ever been considered, no.

KW: No, I mean, not that it should be, but why is it not a consideration?

ID15: Er, well it's treatable isn't it by diet and drugs. It's not something that is incurable, if you know what I mean, it's a gradual thing. It's not just, as an example, Huntington's chorea or something

like that. It's not, you know, where you definitely don't want to have children because of that particular disease.

KW: Yes sure.

ID15: No I don't think it's that serious because it's reasonably treatable.

Again, in the following extract, the interviewee was at pains to distinguish FH from other genetic conditions for which reproductive decision-making would be conceivable. It is noticeable that she was unable to specify examples of what kind of conditions or abnormalities would merit this. This does not however, detract from the strength of her assertions that FH does not fit into this (unspecific) category:

KW: I suppose I was wondering whether it was something you might think about in terms of, well should I have children or should I not have children? That was never a question?

ID60: Oh no, no, no. It's a serious condition, but it's not that serious [...] it's not that kind of genetics where you're thinking, if I was tested would it sway me one way or another, because you know it isn't that much of a problem, in so much as it can be managed. So that absolutely would not be on the table, it wouldn't even be thought about. Whereas people that inherit diseases, they know they've got a gene and it could be passed on, I can't think of anything at the moment, that would cause some other kind of abnormality, then that's different, but it's not even on the continuum I would say.

This distinguishing between FH and other more serious genetic conditions fits with the construction of FH as being part of the normal spectrum of life and illness. Indeed, four of the interviewees drew on ideas concerning normality to show that having FH does not set one apart from one's contemporaries. As one young man put it:

ID112: Other families have different illnesses and different complaints or what have you, so it's just another one on the list.

The previous chapter suggested that FH was not seen as stigmatising and this can be linked to the construction of FH as treatable and, perhaps, as part of the ordinary spectrum of disease. Two younger men discussed this quite frankly,

making a clear link between their (normal) social status and the treatability of their FH, as the following extract illustrates:

ID77: Bear in mind I am on the statins. I'm a normal person now. You know, I'm not, it's a genetic defect, but I've been brought into society, you know what I mean? I'm being brought back to being a normal person.

These ideas about normality were explicitly linked to reproductive decision making in the following example. Here the interviewee explains his rejection of such decision-making in terms of the ability to lead a *normal life*:

ID54: we took 'em to see a consultant paediatrician and funnily enough that was the only negative reaction I've ever had from any medical staff to the condition and he actually said were we, had we ever considered not having children and I was a bit annoyed with him and I said I couldn't understand why we'd take that point of view [...] I mean I was forty-ish then and thought well you know I'd had forty years of normal life with no side effects, even taking the tablets and, you know, why should we not have children, you know.

These data illustrate that rejection of reproductive decision making was tied up with the construction of FH as treatable, manageable and not that serious, and perhaps part of what can be considered normal illness. On the other hand, they also support the suggestion that it is possible to consider FH in terms of reproductive decisions. Two cases have now been noted i.e. one of the clinicians involved with HEART UK and, according to ID54's account, this paediatrician. The patients with FH universally rejected the idea of reproductive decision-making concerning FH, with one exception. In contrast to other interviews, in this case the discussion was generated by the interviewees, a woman and her husband (annotated as H). I had not anticipated that my initial question, about why the interviewee thought she had high cholesterol, would lead onto this talk about reproductive decisions:

KW: So can I ask you what sort of ideas you've got yourselves about why you've got this high cholesterol and why you've had the heart problems? What would you say?

ID101: Oh I think it's just one of those things that happens through family like anything else really. You know families can get things that they pass on, you know, illness.

H101: I suspect she, I'd probably say that if she'd've known when she was younger, before she had children that she had this problem she says she would never of had the

ID101: I don't think I'd've had them, you know

KW: really

ID101: No because me son's ill with it you know and he complains you see

H101: And the eldest girl has got a ???

ID101: Yes, me eldest girl, she's having problems you know with it, so

H101: She's being treated for cholesterol related problems. She's had a minor stroke, it hasn't left her sort of

ID101: deformed in anyway, but she has had a minor stroke, yes.

Compared with the other interviews, the forthrightness of this discussion about choosing not to have children was very striking. Perhaps what was exceptional about this woman's circumstances was the severity of the effect of the condition in her family. Like many interviewees, she reported that a number of people in her father's and her own generation, including herself, had CHD or had died early. More exceptionally, she observed that two of her own children had experienced symptoms related to FH. Witnessing her children's illnesses was pivotal to these discussions, as she put it:

ID101: 'Cos I don't like to think of them being ill, obviously, or, you know, anything wrong with them.

In sum, this section has suggested that, almost without exception, interviewees constructed FH as not like other genetic diseases because it is treatable and manageable. In this way, a boundary was drawn between it and the 'serious' genetic conditions. The data demonstrate that the archetypal genetic condition is serious, untreatable and can legitimately be the subject of reproductive decisions. There was resistance for FH, a treatable, and therefore less serious

condition, to be placed within this category. In one sense, this could be seen as resisting the genetic construction of FH, or at least resisting any challenge to the status of FH as situated within the realm of everyday normal illness. These data support Lambert & Rose's (1996: 79) findings that people with hereditary lipid disorders see their conditions within 'the normality of human imperfection'.

#### **7.4 PASSING IT ON**

Although the idea of reproductive decision making was almost universally rejected, around a quarter of interviewees talked of feelings of regret at the possibility of passing on FH to their children. Just one interviewee expressed feelings of guilt:

KW: So can you remember what you thought at the time, when the kids were being tested?

ID19: I felt guilty when it, well at the time I was worried, I was hoping they didn't have it and I felt guilty that it was through me that they had to come [...] there's no bad feelings [from daughters] you know, that you've passed this onto me now, but you do feel a bit guilty for passing, for having been the person to cause them to have to, because it's not pleasant to have to think about what they're eating all the time.

While this interviewee talked about her own feelings of guilt about passing on FH, she did not blame her own father for passing on the condition. Indeed, there was not a single case where blame was attributed to a parent. In this case, her father passing on FH was constructed as just one of those (unavoidable) things that happen:

ID19: But it's never been any, I mean I don't think about me father you know, oh what's he done passing this, it's nothing, it's just something that happens.

There were a few further cases in which there was an out and out rejection of blame for passing on FH. In the following excerpt, for example, the interviewee rejected blame of his parents or himself because of the limited impact of FH and, again, because passing it on was an (unavoidable) chance thing:

KW: I'm wondering what ideas do you have about why you got FH?  
ID67: It's just the cards I was dealt. That's it. You know, that's the way it is. It's like, I'm about to have a child. The FH hasn't affected that, you know, it's a gamble. The baby may or may not have it and, I mean it's not a debilitating condition. You know I'm perfectly healthy as I am now, and there's treatment for it, so I don't apportion any kind of blame or any guilt or anything like that. It's just one of those things.

Again, FH is constructed as *just one of those things*, as well as a manageable condition, which is treatable, *not debilitating* and compatible with being *perfectly healthy*. The combination of these discourses of FH as just one of those things, that one can not do anything about it, and as a manageable condition, reinforce the construction of FH as part of normal, acceptable, unavoidable illness. It is possible that this construction of FH as highly treatable, unavoidable and unproblematic, function to lay off responsibility for passing on the condition to one's offspring.

The construction of the hereditary aspect as uncertain or contested could also function to lay off responsibility in a similar way. This came to the fore in one very tangible example, in which a man talked in a mixed way about the origins of his son's raised cholesterol. At various points in the interview this was attributed to genetics, but the genetic origin was also contested. The following passages seem to illustrate that the interviewee was struggling to come to terms



with the hereditary aspects of FH precisely because this implies responsibility or blame for his son's raised cholesterol:

(Paras 220-224).

ID13 my son has just gone on medication, he's twenty-one, but he's super fit [...] and we find that hard to take that he's got high cholesterol and has probably never eaten as much as a bag of chips in his life [...] and that sometimes makes me think how important the diet is when you've got this high cholesterol, this genetic type one [...] So that concerns me a bit that he's got the, looks as though he's got the genetic side of it.

(From Para 254)

ID13: nobody is a hundred per cent, even [consultant] and people like that. My son thinks [...] he's convinced that he's inherited it now off me and even we don't know whether it is inherited fully quite yet do we from my experience with the research I've seen, but going back to my point, if they did a gene test and they could say "Oh you've got half a gene or a gene too many," we could actually say we know probably. It's just that you'd worry about whether you've given it to your children or passed it on.

KW: Yes but I mean what do you think, what's your gut feeling, do you think your son has inherited it off you or not?

ID13: No I'm not convinced, I'm not convinced it's inherited yet.

KW: No, but if I put you on the spot and said well okay why do you think your son has got slightly high cholesterol, how would you explain that?

ID13: Well that's a funny one that with [son] because with him being super fit you see he's probably inherited it off me yes (laughs).

It should be noted that these discussions about passing on FH were a relatively minor part of interviewees' talk about having FH. For example, just five people mentioned this topic as the worst aspect of FH. This compared with larger numbers of references to aspects of treatment/medication, the possibility of illness or early death, and changes to diet. In sum, although there was some discussion of regret concerning passing on problems to children, and interviewees' talk appeared to be structured so as to provide a defence against potential accusations of blame, the data suggest that this aspect of FH is not at the forefront of patients' constructions of the condition.

## 7.5 LIVING AT RISK OF CHD

Interviewees' almost overwhelming rejection of reproductive decision making has already been discussed. Novas and Rose (Novas & Rose, 2000; Rose, N., 2001; Rose & Novas, 2004) notion of genetic prudence suggests that having knowledge of genetic risk information is associated with obligations regarding other areas of life planning such as financial and occupational planning. There was very little talk concerning the influence of FH on life plans in these interviews. Just three interviewees made any kind of reference to knowledge of being *at risk* of CHD having influenced their life plans, as distinct from talk of changes made as a result of actually having CHD, and in only one case was this linked directly to responsibilities to other people. Here the interviewee said that one of the impacts of having FH was that he had chosen a job with a stable 'blue chip' company with 'a good pension' to make sure that his wife and family would be financially secure should he experience health problems. In the other two cases the interviewees talked of making sure they did things they wanted to do, or that the diagnosis had made them think more about what they wanted to do. In short, although ideas about life planning were present, this was hardly a major theme in the interviews.

The previous chapter and the first section of this chapter suggest that FH is constructed as treatable, manageable, unavoidable and relatively unproblematic, and that CHD is seen as largely avoidable by taking care of oneself by maintaining appropriate lifestyle, taking medication and following medical advice. So far the analysis has suggested that these ways of framing FH and CHD underlie interviewees' constructions of responsibilities to self and

discussions concerning having children. While talk of the treatability and manageability was very prominent, FH was not constructed as entirely unproblematic. At least a third of the interviewees said that they thought of FH as a serious condition, relating this to early deaths of family members or talking of its potential to cause CHD, and several others said it would be serious if it was not controlled or treated.

The idea that predictive health risk information might result in a new health status between health and illness has been prominent in scholarly discussion (inter alia Armstrong, 1995; Crawford, 1980; Davison, 1996; Finkler, 2001; Scott et al., 2005). Four of the interviewees talked about being or having been alert to particular bodily feelings. These were sensations in the chest which were associated with anticipating heart problems:

ID67: it's always in the back of your mind, if you ever have a twinge in your chest you sort of, at my age, you start thinking of well what's that? Is that anything to do with that?

In one other case, a woman talked about FH as being frightening and something that was 'in your head all the time'. These expressions of bodily awareness or constant consciousness are perhaps manifestations of the liminal status (Scott et al., 2005) of people living 'at risk' of CHD. This was, however, a relatively minor theme in interviewees' talk about their experiences of FH.

A second theme concerned the resistance of younger people, in particular, to engaging with predictive health information of this nature. Around a third of interviewees made some reference to the relevance of age and life course. Ill-health was described as something that young people do not, or do not want to,

think about. Interviewees talked about the difficulty of persuading their adult children to be tested or attend the clinic, and of their own lack of engagement in earlier years, because ‘when you’re young you think you’ll live for ever’. A handful of the interviewees was diagnosed with raised cholesterol in their teens and early twenties, and three of these suggested that this information had been actively rejected or ignored in earlier periods of their lives. Two of these, both women and diagnosed at the ages of sixteen and twenty-four, framed their diagnoses almost as *unwanted* information; that an awareness of their own mortality had been prematurely imposed upon them:

ID60: You see most people don’t think about heart problems I should imagine until they get older. Whereas I was thinking about it from the age of seventeen, eighteen, and really as a woman and the menopause [...] you really can get to forty odd and it’s not a problem. Whereas I was thinking it was, because it was told to me. People don’t think about when they’re going to die. Whereas I started thinking about it literally from the age of, I would say, seventeen.

ID97: At the time I didn’t want to know about it. At the time I wanted to know about it after menopause when things became more important to get things sorted out. I thought nothing can go wrong with me; I’m only twenty four.

The notion that there are certain periods of life that are, or should be, free of thoughts of death and illness and other periods associated with increasing health-consciousness were present in several other interviews. In the main, however, talk of worry or of resistance to knowledge of one’s at-risk status were not prominent aspects of patients’ discussions about FH. The framing of this predictive health risk information as largely unproblematic is particularly significant in the interviewees’ talk about their own children, which is discussed in the next section.

## 7.6 TAKING CARE OF CHILDREN WITH FH

In a previous section it was noted that, although interviewees sometimes expressed regret for ‘passing on’ FH to their offspring, this constituted only a minor theme in their talk about having the condition. By contrast, interviewees talked much more about their role in the care and treatment of their existing children. This was evident in discussion about taking children, or persuading/encouraging adult offspring to attend for cholesterol testing in order to establish a diagnosis of FH, and encouraging appropriate behaviours and compliance with medication.

Because I initially understood FH to be an adult-onset condition, it took me quite a while to recognise this area as an important theme for the interviewees. In order to understand this section, it is worth outlining recommendations concerning the diagnosis and management of FH in children. Clinical guidelines produced by the BHA (Wray et al., 1996) recommend that children should be screened for FH between the ages of two and ten, with dietary modifications encouraged from the age of two. Recommendations concerning the type of medication to prescribe and the age at which it is appropriate to start prescribing seem to be more equivocal (Durrington, 2003; Greene & Durrington, 2004; Marks et al., 2000; Wray et al., 1996). It is, therefore, likely that parents may be encouraged to have their children tested from an early age, although advice on treatment and management may vary<sup>17</sup>.

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<sup>17</sup> Recommendations about FH are consistent with national guidance concerning genetic testing of children for adult onset conditions (Advisory Committee on Genetic Testing, 1998). This precludes only testing of young children where therapies are not available. Although there is no discussion of any examples such as FH, in which treatments/behavioural regimes in childhood may help to prevent disease in later life, the guidance provides no obstacle to the early screening of FH.

More than half of the interviewees talked at some point about taking children or having been taken as children for testing. This was, by and large, constructed as wholly obvious, straightforward and unproblematic; it was not framed as an action that would reveal something about one's inner-self or identity, that might cause anxiety, or that might bring children prematurely into the medical sphere, but rather as a very practical act. ID11, for example, discussed taking her young daughter for testing. Her talk indicated that this was not something that was a matter for choice, but an obligation (*she's got to have it*), and that it was a practical and immediate matter that needed to be *sorted out*:

ID11: I was advised to have her tested when she was two, so she's had tests since two.

KW: Right, and I don't know if this is a relevant question, but I just wondered what kind of decision that was whether to get her tested or not. Was it obvious or was it difficult?

ID11: Oh yes, no, it wasn't difficult because obviously mine was very high when I first and no, it wasn't difficult. It was just something that you know, I thought, well, she's got to have it, if she's got it, it's best to get it sorted out.

In the following extract a woman and her husband (denoted as H) discussed asking their teenage children to go for testing:

KW: You said we asked for our boys to be done and they got tested as well. Can you just talk me through how that worked?

H33: Well, it just, I mean, we didn't insist on them, but we told them the situation with [wife] and that they should be tested, simple as that, and they did, didn't they?

The use of the phrase *simple as that* is particularly striking here. It again suggests that the diagnosis of children is uncomplicated for both parents and children. In the previous section it was suggested that interviewees who had

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been diagnosed at a young age occasionally talked of their anxiety or resistance. This was particularly so in the case of ID60, who was diagnosed at the age of sixteen. She talked of being in ‘complete denial’ about the diagnosis in her earlier years, that she ‘completely ignored it, but at the same time had real psychological problems in terms of thinking I was going to die early’. Yet, even this woman constructed the testing of her own children as obvious and unproblematic, akin to taking them for childhood vaccination. She said that her two children were tested at the age of six months and in the following extract she explained how that came about:

ID60: I think it may have been my dad, because I went back to work straight away with [daughter] [...] My dad tended to do most of the running around for me. So I think he did the mumps and measles and all the other stuff [...] I can’t recall actually taking her myself, so it must have been my dad. Which I know that she was tested.

KW: I’m just wondering whether that was an obvious decision or whether that was something you had to kick around with your husband?

ID60: No that was, it wouldn’t have been anything that would’ve been discussed in depth, because there might be two outcomes. It was just anything like that, I just make the decisions on anyway. So, no, that was just an obvious choice.

She manages the disparity between her own experience and her actions toward her children by drawing a distinction between them, describing the situation with her children as ‘a different scenario altogether’. This is achieved by drawing on parental familiarity with the issues and parental responsibility. The diagnosis is constructed as problematic only under certain circumstances, where there is no ‘parental backup and knowledge’, a situation that would require additional thought and support. In this way she demonstrated how she has acted responsibly towards her children, firstly by making sure that they were tested and secondly by showing how this decision took into consideration their psychological welfare:

ID60: Had either of them got raised cholesterol levels I would have been able to have dealt with it and therefore they would have dealt with it. So that's a different scenario altogether. Children that are diagnosed early, I think, as long as they have the backup from parents, maybe with a large percentage of cases, it's probably because parents have got cholesterol anyway. So I think that if the parents do, that would be fine. If they hadn't, I think there is a case for actually doing quite a lot with children if they haven't got parental backup and knowledge, just to deal with it, because you don't know what people are thinking in their internal life and, actually, it can be a big problem, I think.

This extract indicates that parents have a duty to provide 'backup' to their children. Only one other woman hinted at any psychological aspects to her parenting role, of 'trying to train them to look after themselves' without causing them anxiety, 'without making them feel that they've not got a full lifespan ahead of them'. For the other interviewees, talk of their actions relating to their children focussed on being mindful of their children's diets, and of encouraging or training their children to take their medication. In the following extracts, for example, the interviewee talks of getting his children into the *habit* of taking their medication and of having to *give some consideration* to his children's diet:

(From para 85)

ID54: I wanted to make sure that all the family, the kids, if they needed the treatment they got it straight away and we make sure that they take their medication and I would think that they're probably alright and live a normal life.

KW: How do you make sure that they take their medication?

ID54: Er well, have you taken your medicine yet? Before you go to bed. Yeah

KW: okay

ID54: It's because we've started them at a young age, it's a habit.

(Para 153)

ID54: I mean my children have gorrit and I don't see how it will affect their lives at all. Dietary, I mean you've got to give some consideration to not taking the children to MacDonald's everyday for their meals and you know we tend to eat lots of white meat, chicken, turkey, tuna, very little red meat at all. We're not great cheese eaters so.



It is also noticeable in these extracts that the interviewee connects his care of his children with the idea that they will probably have a *normal* life. This suggests that demonstrating responsibility for managing one's children's cholesterol, as well as constructing this as a largely pragmatic and unproblematic area, may be connected to warding off reproach for 'passing on' the condition in the first place.

There was only one case in which an interviewee talked of declining to have her child tested. ID100 drew on a discourse of protecting her son from early diagnosis because she wanted him to be able to be a child with 'no labels' attached. Her talk makes very clear that the fact her son has not been tested is not a matter of ignorance or fecklessness; this was a measured decision based on his psychological welfare and contingent upon his maintenance of appropriate behavioural regimes:

ID100: They did ask me, I've been asked by several people. I feel he's ten years old, he's a fit child. I know he's male, so his risks are higher if he's got it. He's very active. He eats the diet that I eat and I just feel now he's a child and I want him to be that with no label attached. I don't want him to have a label. And if he follows that regime for now, okay.

She also drew on the notion of 'choice' concerning testing, an idea more familiar from the predominant discourses about the application of genetic technologies. This reinforced the construction of her decision as measured and ethical:

ID100: For adults, yes, I think once you have the right to make your own choice if you go for that test, if you 'ave a symptom or if you're worried, that's your choice. But you shouldn't inflict, I feel on your child. It's got to be his choice when he's old enough to make the choice.

The effect of this account was to present the interviewee as a considered and responsible parent; parental duties were demonstrably discharged through the provision of appropriate behavioural regimes, consideration of the children's psychological welfare and appeals to autonomy in testing decisions.

These data on testing and diagnosing children illustrate that this can be constructed as troubling information or an unnecessary intervention. It was, however, mostly constructed as untroubling and the analysis suggests that, in these cases, moral probity was demonstrated through talk about having one's children tested and their continued care. In other words, whether children have been tested or not, these accounts position parents as responsible in one way or another.

Parental responsibilities, evidently, continued into adulthood. A number of interviewees talked of trying to persuade or encourage their adult offspring to get tested, to maintain healthy behavioural regimes and to take their medication, or of their parents doing this to them. ID101 and her husband framed themselves as responsible not just for their own children, but also their grandchildren. For example in the following extract they talk about trying to persuade their twenty-one year old grandson to have his cholesterol tested and to change his behaviours:

ID101: He doesn't really want to know, but we keep at him

KW: What do you say?

ID101: You must go, you must go, you know, eventually he will.

H101: Well, we badger him about cut down on your smoking and what have you.

The welfare of one's offspring is an unavoidably moral area (see Murphy, 2003; Ribbens McCarthy et al., 2000). The data presented suggest that interviewees may take different approaches to discharging their duties to their offspring, but however this is achieved it is difficult to remain silent on this topic. Indeed all but three interviewees contributed to these discourses concerning the diagnosis of children or the care of adult offspring and two of these did not have children. There was only one case where an interviewee was silent on the testing or welfare of his children. In the following extract it is notable that the interviewee is silent about his two older children. There is no attempt to confirm that he has told them about FH, or, alternatively, to justify why he has not told them. This could be framed as a rejection of obligation or, perhaps, an absence of obligation based on genetic connections:

KW: And can you remember when you got the diagnosis, who did you tell about it? Did you talk about it within your family?

ID35: Yeah, my wife came with me

KW: And what about your kids? Have you got kids? Sorry.

ID35: Yeah

KW: How many kids have you got?

ID35: Two that I don't see at all from a previous marriage and one that I see regularly, but she was only three at the time

KW: And have you talked to them about it now?

ID35: No, she's not mine actually, she's me step-daughter, so it's not familial, I'm not passing anything onto her.

KW: And have you talked about it in your wider family?

ID35: No we're not very close

What do these data, in sum, say about the genetic construction of FH? The prevalence of talk in these interviews about actions in relation to one's offspring may be an acknowledgement of the familial aspect of the condition and of genetic responsibilities that flow from this. It is, in some ways, hard to separate this from the responsibilities that any parent feels for the continued well-being of an offspring with any health-related issue. One woman explicitly

connected her duties to her children with her responsibility for passing the condition on. In the extract she is talking about her reaction to being asked to have her children tested:

ID18: I thought, that she was telling me like that whatever I'd got passed on to me kids and I didn't like that idea obviously and that's why I made them go. I said I don't want you going through what I've been through and if you listen now at this age and take what you've got to take, perhaps you know you'll avoid having a heart attack or whatever.

This comment helps to support the suggestion that interviewees' talk about their offspring was not just concerned with parental responsibilities, but with genetic responsibilities. In sum this section has suggested that interviewees largely framed the testing and management of their offsprings' cholesterol as an unproblematic and practical issue. Nevertheless, care of offspring was almost without exception constructed as an area of responsibility. It is suggested that these responsibilities flow not just from parental responsibility in general, but also through more specific hereditary linkages.

## **7.7 TALKING WITH KIN**

In their studies concerning hereditary breast/ovarian cancer and familial melanoma, Hallowell (1999) and Polzer et al. (2002) have suggested that participants expressed a strong sense of obligation to communicate genetic risk information with their kin and to encourage them to manage these risks. These studies suggest that this applies particularly to children, but also to siblings and wider kin. These findings are consistent with other studies concerning the communication of genetic risk information within families by people attending for genetic counselling and testing (d'Agincourt-Canning, 2001; Forrest et al., 2003; Green et al., 1997). In the present study, as the previous section has

argued, there was an explicit discourse of responsibility regarding the welfare of one's offspring. By contrast, talk concerned with informing and educating other kin assumed much less prominence. Indeed discussions concerning wider kin proved quite difficult to elicit in some cases. This suggests that family connections are not at the forefront of thoughts about FH.

In terms of the interview process, the interview topic guide included the question 'who did you tell/talk to about the diagnosis'? I had anticipated that this question would prompt a discussion about the communication of information with kin and possibly other people. However, a number of interviewees did not understand the question, or did not understand it in this way. The question, therefore, did not have an obvious logic for the interviewees and details about communicating with kin then emerged at other points in the interview, sometimes after considerable probing by me. It is important to note that, in contrast with the studies cited above, the analysis is not so much concerned with whether interviewees had or had not communicated with their kin and their reasons for this, but with the way this area was presented or not at the interview. This is a concern with the construction of moral meanings; the analysis is concerned with whether interviewees need to demonstrate they have communicated with their kin in order to be seen by themselves and others as proper and responsible people with FH (see Murphy, 2000).

According to the interviewees' accounts of how they came to be diagnosed, there must have been a certain amount of information sharing, since three

interviewees talked of attending for testing at the instigation of a sibling. Talk of communicating with kin mostly involved siblings, but sometimes included wider kin. As with other studies of the communication of genetic information (d'Agincourt-Canning, 2001; Forrest et al., 2003; Green et al., 1997), interviewees talked of informing those with whom they had social or geographically proximity. While other studies have reported that a genetic diagnosis may lead to some tracing of family members where contact has been lost, there was just one example of this suggested in these interviews; a diagnosis of FH is not a reason to contact kin one has fallen out with or does not know.

Interviewees' discussions about communicating with their kin can be typified in four main ways:

1. **Claims responsibility:** those who readily credited themselves as having talked with kin or as being instrumental in others attending for testing.
2. **Not claiming responsibility:** those who talked of telling kin, but did not obviously present this as a purposive action out of concern for their welfare. These interviewees' talk of communicating with kin sometimes emerged only through persistent questioning on my part.
3. **Not telling:** those who did not appear to have talked with their kin.
4. **Refuting responsibility:** those who actively refuted responsibility for talking with kin.

Only four interviewees clearly framed themselves as having a role in telling their kin about the condition and persuading them to be tested. ID24 can be

seen to claim responsibility for, or attribute himself a central role in, the welfare of his kin, including his siblings and cousins. This is established particularly though using the first person to describe events, *I talked to them, I wanted them to come, I couldn't convince them, I made sure they all know:*

KW: After you saw [clinician] what did you do then, I mean who did you talk to about the diagnosis?

ID24: Well I talked to m' family initially, cos I wanted all of them, I wanted them all to come down, I wanted them all to get involved, but I couldn't convince them all at the time. I mean I've got three children [talks about children]. I've got a twin sister, who actually did go, she's the only one who went, she was OK [...] My older sister has been, has found she's got a high cholesterol problem and I said to her well, ask your doctor for a referral and she's not been [...] I mean I said, well I wish you get onto the clinic here, I'm sure you could if you asked for it. But she's not pushed for it yet [...] But I said, well you should be on these books here I think, but anyway.

[more conversation about children and family history]

KW: But not the wider family, you've chatted within your own immediate family?

ID24: Well me own immediate family, yeah, I mean I've got cousins as well. I've got a cousin who also I think might have the same problem and again you know I've told her about it [...] I made sure they all know, because I want them to be aware of it, cos it could affect them.

The majority of interviewees who mentioned talking with their kin could be described as not claiming responsibility. This tended to involve less directive language. In the following example, the interviewee answered my question about who she told by talking about which of her siblings have raised cholesterol. This demonstrates that she had taken an interest in their cholesterol status i.e. it was a relevant subject. However, her use of the passive voice in talking about their testing, *they were advised to have a cholesterol check, I was told to tell relations*, suggests that she was reluctant to claim the credit for this; the responsibility for getting kin to come for testing was mediated by the clinic:

KW: When they told you it was this, some kind of family thing, can you remember whether you told anybody? I mean, I'm not sure if you've got siblings or anything?

ID11: Yes my brother has suffered with high cholesterol. I have two sisters. They don't.

KW: Yes and do you know how your brother and sisters came to find out about it? Who found out first say? You did?

ID11: Yes and then they were advised to have a cholesterol check and as I say my brother has high cholesterol. But he's also had a by-pass 30 odd years ago so

KW: Okay, I was just wondering whether, you said they were advised to have tests. I'm wondering how, whether you were the conduit, you were the one that said go?

ID11: Yes, yes I was told to tell, you know, any relations, well brothers and sisters, to have a blood test.

In another example, the interviewee's talk suggests that he had a certain amount of discussion with his siblings about the raised cholesterol condition, but this was not framed as particularly urgent or significant. The extract illustrates the relevance of social proximity to discourses about communicating with kin, through the reference to *only meeting at weddings and funerals*. The interviewee's discussions with his siblings were framed as more to do with accounting for his own CHD than concern for their welfare. It is through my persistent and rather single-minded questioning that he reiterates that he did speak to his siblings about the hereditary aspect of the condition. It is notable that it is my construction, not his, that his sister attended for tests *as a result of him talking to her*:

KW: And so did you ever talk to them [brothers and sister] about it?

ID13: We did do, yeah, a long while ago but with marriage and children and things like that we only meet at weddings and funerals

KW: Right, right.

ID13: Yeah so I don't know how they're going on. I think my sister attended, not here but I think she had a slight problem with cholesterol at one point.

KW: But were you the first person to be told that it was this family thing?

ID13: Yes, within the family I was the first one.

KW: So were you the one that had to tell the others or had they found out through another way?



ID13: I don't think it was really, it wasn't really mentioned too much until I went in hospital and had an operation, had one of the operations and they said what's up with him, oh it's all to do with his high cholesterol stuff and basically like that really. But I did speak to them [...] I'm almost sure my sister had some tests done at her local GP where she lived.

KW: as a result of you talking to her about it, yeah?

ID13: Yeah, just word of mouth really you know saying it could be inherited and stuff like that.

This was not the only interview where talk of discussing the condition with kin was related first to the welfare of the interviewee rather than the welfare of the kin. Interviewees also sometimes said they did not know how their siblings had come to be tested or whether they had been tested. These data suggest that communicating with kin about FH out of concern for the welfare of these kin is not always seen as an imperative. Although many said they had talked with their kin, there was not an explicit discourse of obligation.

In a small number of cases, people did not appear to have talked with their kin. ID04 provides an example of someone who did not understand my question and who had, apparently, not talked with her kin. Immediately preceding this extract we had discussed the clinic appointment at which the interviewee was diagnosed. She described this as involving a discussion of her family background, including 'me mum and me dad and me brothers and sisters'. This establishes that the interviewee has siblings and they are somehow relevant to her own diagnosis, but her talk is silent on whether she communicated information with them, suggesting a lack of obligation about the condition:

KW: So when you first were told you've got this high cholesterol problem can you remember who you told about it?

ID04: What d'you mean, who I told?

KW: Um like in your family or at work maybe?

ID04: Um me partner, when I went home, me husband.

KW: Right, right, and was there anybody else you told at all?

ID04: No

In two cases the responsibility for telling kin implied by my questioning was plainly refuted. In both of these cases not telling was attributed to social and geographical distance. This, in itself, was not unusual, as other people also talked of relatives they had not communicated with because they were not close to or in touch with them. It was the provision of explicit or further justification that made these cases distinctive. These justifications suggest that the interviewees recognise that communicating with kin could be constructed as an obligation and that their actions are possibly being called into question. It is this recognition of obligation implied within these accounts that sets them apart from the other interviews.

In the first of these, the interviewee described his family as ‘very loosely knit’ and explained that his brothers lived abroad and that he had sporadic contact with them, through ‘Christmas cards’ and ‘a ding on m’ mobile’ a few times a year. In the following extract he talks about whether he has been in touch with his brothers about the condition. It is notable that he immediately recognises the issue that I am trying to address with my questioning (*I can see where you’re leading*) and his response is quite defensive. This suggests that he recognises the implied responsibility. It is also notable that he does not confirm or disconfirm whether he has tried to talk with his brothers in the past, although his talk implies that he may have tried (*its something they clam up on*). Finally, his justification for not telling his brothers is in terms of their attitude, advice about getting tested would be rejected (*mind your own*

*business*), and he makes plain that getting tested is a matter of individual responsibility (*if he wants to get checked, he will*):

KW: So you never really talk to them about it, or told them about it.

ID35: Oh no I wouldn't anyway, no

KW: And you wouldn't know if they've got any heart problems or cholesterol problems?

ID35: I can see where you're leading

KW: Where am I, tell me where I'm leading (laughs)?

ID35: To see if it's familial, see if we've discussed it, and see if they've got it. It's on the maternal side. My mother died of heart problems, had an uncle, he died at thirty nine, when they knew nothing about this, with a heart attack. So I'm not sure if they've been checked or not, me brothers, because it's something they clam up on. If I phoned [brother] and said, have you been checked for cholesterol levels, he'd probably say, 'it's red hot here', you know mind your own business, don't bother, we know each other. If he wants to get checked for 'em he'll get checked for 'em without me as, so we don't discuss it, no.

In the second case, the interviewee reported that he had not discussed the condition with his brother because of both social and geographical distance (*they don't get on*, and they live in different parts of the country). This talk is then connected to a discussion about the health status of his father and uncles. The significance of this talk is not immediately clear, but implies that the raised cholesterol is sporadic and has unknown outcomes. After all, one of his uncles has lived successfully with angina for fifty years and is now a ripe old age. The function of this talk may be to individualise the condition, detracting from its familial aspects and therefore mitigating his lack of contact with his own brother. Earlier in this chapter, it was suggested that the construction of the hereditary aspect as uncertain or contested may function to lay off responsibility for passing on the condition to one's children. It is possible that this man's talk functions in a similar way to lay off responsibility concerning his brother:

KW: I was just wondering whether there'd been any kind of discussion with your brother?

ID88: no, we don't particularly speak, you know, we don't get on terribly well. He lives in London so, and I live in [town]. But you see if you want you know, just as a note, my father died of a heart attack in 1969, so did his brother, okay. Now here's the twist in this. His other brother is ninety four next month and has had angina for fifty years and never had an operation, and he's still playing the accordion, the keyboard and a successful artist, now put that in your pipe and smoke it, init really? And his other brother is about eighty nine, so I dunno. There's no answer is there.

To sum up, this section has suggested that only a small proportion of interviewees actively constructed talking with kin and encouraging them to manage their cholesterol as an obligation. Although many of them may have talked with their siblings or other family members about FH or raised cholesterol and know the status of these kin, family connections were not a prominent part of their construction of FH. Occasionally, obligations based on such connections were actively rejected.

## **7.8 TALKING WITH OTHER PEOPLE**

The previous sections have focussed on obligations regarding kin. This section is concerned with wider obligations to other people. More than a third of interviewees talked of discussing cholesterol or FH with their friends, colleagues or other people in order to draw attention to these conditions and encourage these people to get tested or change their lifestyles. These interviewees could, perhaps, be described as constructing themselves a role as cholesterol ambassador or champion. In the minority of these cases (4), this educational role linked directly with the possibility that others may have FH or an hereditary form of raised cholesterol. One woman, for example, reported that she had encouraged a colleague to get his cholesterol checked, because

both his parents had died of heart problems at a young age. In the main, however, this educational role seemed to be less specifically concerned with FH or hereditary forms of raised cholesterol, and more about recognising and managing raised cholesterol in general. This suggests that interviewees have a responsibility to others based on their more general knowledge and expertise concerning cholesterol problems and management. A few people (3) talked of being prompted to talk with others because they recognised that these people had some visual signs of a cholesterol problem, such as deposits around the eyes, as in the following example:

ID19: I have told people, that I've met that have had these things. Three people up to now, that I've seen with these things and spoken to them about it and explained, that I had those and I'd got rid of them with medication because I have high blood fats, which can be quite dangerous.

There were other occasions where interviewees reported undertaking more general awareness raising, seemingly unprompted by indications of any specific problems. The following excerpt stands out as being a very clear example of cholesterol championing:

KW: Do you think anything good's come out of it [the high cholesterol problem]?

ID85: Well I spread the word more than what I would've done before. I think I might have influenced half a dozen people to have their blood checked for cholesterol and with my job [joiner] I don't work sites, I work customers, and most of the time we get on the friendly path and I would say fifty per cent of the time cholesterol may come into it, you know, I would suggest to them to go and have your blood tested, 'cos it doesn't matter how old you are, it is important.

These data suggest that responsibilities to others are not solely derived through genetic connections, but a more general duty to spread the knowledge one has and help others where possible. Hallowell's (1999) study concerning genetic responsibility also considers the expression of wider obligations and suggests

that responsibilities to kin and to others may be derived as much through social as through genetic connections. Hallowell's suggestion is supported by the data presented in this and the previous sections. These data also suggest that even though interviewees may distinguish between their own hypercholesterolaemia and other people's on aetiological grounds, as illustrated in the previous chapter, they do not necessarily maintain this distinction in terms of their responsibilities to raise awareness and encourage action. This contributes to the sense that FH is constructed as a cholesterol problem as much, if not more than, as a hereditary problem.

## **7.9 CHAPTER SUMMARY AND DISCUSSION**

This chapter has argued that one marker of the genetic construction of a condition is the degree to which obligations to others are derived through genetic connections. The analysis suggests that the idea of reproductive decision making in relation to FH was almost universally rejected, setting it apart from other genetic conditions which were seen as more serious.

Discussion of other areas of life planning in relation to FH were largely absent. The rejection or absence of discussion of life planning can be attributed to the construction of FH as unavoidable, manageable and largely unproblematic.

Interviewees expressed a strong sense of obligation concerning the welfare of their offspring, to make sure they are tested and encourage them to manage their cholesterol. While it is difficult to distinguish the extent to which these obligations to offspring flow from parental responsibilities in general or from genetic linkages more specifically, it is suggested that they can at least be partly attributed to genetic responsibility. Obligations concerning other kin were much less clearly defined. Only a small proportion actively claimed

responsibility for communicating information to their siblings and other family members, and for encouraging them to get tested and manage their cholesterol. Furthermore, more than a third of the interviewees constructed an area of responsibility for communicating information about cholesterol and FH and encouraging other people in general to get tested. This confirms that responsibilities to others are not solely derived through genetic connections, but also through social connections. In sum interviewees' constructions of their responsibilities are not strongly framed through genetic connections, which suggests that FH is not predominantly seen through a genetic lens.

In relation to the geneticisation thesis, these data reinforce the findings of the previous chapter; genetics does not appear to be the dominant discourse in interviewees' accounts of their actions in relation to FH. Interviewees framed FH as part of normal acceptable illness. In other words, the contribution of an hereditary aetiology to FH does not set it apart from other kinds of illnesses. Interviewees resisted any challenges to their ideas about normality and abnormality. In the previous chapter it was suggested that interviewees' accounts of the causes of hypercholesterolaemia distinguished people along genetic lines. The data here suggest, nevertheless, that a proportion of interviewees do not maintain this distinction when it comes to talking with other people to raise awareness and encourage action in relation to cholesterol, blurring the boundary set up in their talk about aetiology.

## CHAPTER 8: CONCLUSION

This thesis set out to examine the geneticisation thesis by exploring lay and professional constructions of CHD and FH, focussing particularly on the work and talk of HEART UK and publications of its professional members, and on the views of ‘ordinary’ patients with FH. Chapter 3 argued that the geneticisation thesis makes a set of claims about the changing place of genetic knowledge in concepts of health and illness and the impact of this knowledge on health care practices, and values and attitudes. It went on to suggest that each of these claims might be explored empirically. The aim of this thesis was to focus on these empirical questions about whether and how genetic knowledge are changing disease concepts, health care practices and values and attitudes in the case of CHD and FH. It is one of very few empirical studies of geneticisation and even fewer studies concerned with genetic constructions of complex common conditions that are managed outside of clinical genetic services. Chapter 3 also suggested that empirical enquiry concerning geneticisation may consider data on a number of different levels and this study has looked at constructions of CHD and FH across several domains, including biomedical literature, clinical discourses and patient discourses. The thesis also aimed to contribute to discussions about the influence of lay health groups, particularly those associated with genetics, on the production and dissemination of biomedical knowledge. In sum, the data challenge the geneticisation thesis in a number of important ways. They also suggest that HEART UK has not adopted the radical practices with regard to lay and expert relations reported by other studies.



This final chapter will summarise the main findings of the thesis and will then discuss these in relation to ideas about expertise, the notion of genetic responsibility, and the geneticisation thesis. It then considers the implications of the findings for policy and practice and concludes by discussing further questions that are raised by the findings.

## **8.1 SUMMARY OF FINDINGS**

The thesis explores geneticisation by asking

*to what extent do FH patients and biomedical professionals construct FH and CHD as genetic conditions?*

This question was divided into three further questions

(1) *How are FH and CHD constructed in the recent biomedical literature?*

(2) *How does HEART UK construct FH and CHD?*

(3) *How do patients with FH construct FH and CHD?*

The thesis also aimed to explore how HEART UK's activities and discourses should be understood, meaning the sense in which these can be thought of as indicative of lay constructions of FH and CHD. The thesis therefore asked a fourth research question

(4) *What roles do patients play in the constructions of discourses about FH and CHD within HEART UK?*

This section summarises the main findings concerning these four questions.

### **How are FH and CHD constructed in the recent biomedical literature?**

The analysis suggests that there are a variety of discourses about CHD within the biomedical literature. The analysis of the four recent commentary papers in CHD presented in Chapter 4 suggested that there is a model of CHD that prioritises the influence of genetic variations on disease susceptibility as illustrated by Stephens & Humphries (2003). However, this represents just one strand of research in the field of CHD. The analysis suggests that there are a number of alternative and competing model, which draw on a range of emerging and established risk factors. Beaglehole & Magnus (2002a), for example, classify multiple gene influences as just one of six emerging risk factor areas for CHD, in addition to the established risk factors. They name the other emerging areas as thrombotic factors & other biochemical markers, inflammation & infectious agents, early life exposures, oestrogen deficiency, and psychosocial influences. It is possible that other researchers in the field of CHD might characterise the emerging risk factors differently and these six areas should not be seen as an exhaustive list. The analysis, nevertheless, highlighted the heterogeneity of current research and models concerning CHD aetiology.

The HEART UK professionals studied had little involvement with genetic models of CHD in general and their interest in genetics was limited mainly to specific hereditary lipid disorders. The research that they undertook in connection with aetiological rather treatment and management issues was mainly focussed on the molecular processes involved in atherosclerosis i.e. lipid, inflammation and clotting pathways. This rarely included analysis of

genetic variations linked to these pathways. In relation to Beaglehole & Magnus's (2002a) analysis, this is an interest in two of the emerging factors i.e. thrombotic factors & the effect of biochemical markers, and the role of inflammation & infectious agents, although here the emphasis was on inflammation not infections. It is notable that this interest in molecular models is not the same as an interest in genetic models that draw on inherited differences.

Even in the case of FH there seemed to be some lack of certainty in biomedical constructions about the role of genes. Analysis of the Simon Broome group publications demonstrated that the current diagnostic criteria for FH draw on a set of clinical indicators concerning blood cholesterol levels, the presence of tendon xanthomata, and a family history of hypercholesterolaemia or early CHD. The group has recently started to discuss the possible benefits of including DNA-based testing into the diagnostic process. Their discussion suggests that models of the genetic basis of FH have become more complex during the last decade or so, due to research involving genetic analysis that has raised questions about the relationship between the presence of an FH mutation and a clinical diagnosis of FH. Although the Simon Broome group publications have constructed genetic testing as an inevitable development contingent largely upon technical advances, one of the group's members, Paul Durrington, has displayed considerable ambivalence in his individual publications about the utility of genetic testing. This suggests that FH cannot simply be defined as a monogenic hereditary disorder caused by a mutation in the LDL receptor or other specified genes. Some clinicians may privilege a

clinical definition based on the presence of hypercholesterolaemia, tendon xanthomata and family history, regardless of the precise genetic aetiology.

Chapter 4 argued that constructions of FH and CHD might fruitfully be analysed along disciplinary lines. The geneticists, epidemiologists, cardiologist, lipidologists and other biomedical professionals discussed in the chapter may have different aims, priorities and ways of working which are reflected in the different models of FH and CHD that they tend to privilege. This idea will be expanded on in following sections of this chapter.

#### **How does HEART UK construct FH and CHD?**

This research question was intended to capture both explicit accounts of FH and CHD in HEART UK's written texts and in the interviews with HEART UK members, and the constructions of CHD and FH embedded in the aims and activities of the organisation. HEART UK's texts present the causes of CHD mainly in terms of the established risk factors, dividing these into non-modifiable, i.e. age, sex and family history, and modifiable, i.e. 'lifestyle' related, categories. Interviewees similarly constructed two main categories of hypercholesterolaemia, hereditary and lifestyle-induced. The hereditary category was limited to the recognised familial lipid disorders. There was practically no discussion of multiple gene influences on susceptibility to CHD or hypercholesterolaemia. The models of these conditions continue to attribute a high degree of personal responsibility to causation and prevention, conforming to established public health discourses in these areas (Davison et al., 1989, 1991, 1992; Petersen & Lupton, 1996).

Following on from the findings concerning constructions of FH in the biomedical literature, there was ambivalence amongst both the professional and lay members of HEART UK about the clinical utility of genetic testing for FH. This again suggests a definition of FH that prioritises clinical attributes such as hypercholesterolemia and family history, over genetic attributes. The data provided further evidence that different views on DNA-based testing may reflect disciplinary differences. The fact that two of the professional members, both clinicians, were largely positive about DNA-testing complicated this issue, suggesting that opinions do not align strictly down disciplinary lines. Nevertheless, the introduction of DNA-based diagnosis for FH did not appear to be prioritised within the organisation.

Chapter 5 showed that HEART UK's aims have broadened from their original focus on the hereditary lipid disorders to include CVD risks in general. This is symbolised in the progression of the organisation from being *the family heart association* to becoming *the cholesterol charity*. The organisation is now involved in a range of activities concerned with FH specifically and CVD risk more generally, although interviewees differed in their views concerning how much the organisation had expanded or should expand its role. It is notable that little in the organisation's work is orientated specifically to the genetic aspects of FH. Its collaborative and advisory activities mainly involved groups involved with CVD, or matters concerning CVD. This orientation was also embedded in the professional membership of the organisation. It could perhaps

be described as being infused with a CVD culture rather than a genetics culture.

### **How do patients with FH construct FH and CHD?**

The patients with FH interviewed at the lipid clinic had multiple and sometimes contradictory ways of constructing the condition. It was not necessarily characterised solely as an hereditary problem, but also as a problem of the liver or as a cholesterol problem. Interviewees constructed the same two categories of hypercholesterolemia found in HEART UK's constructions.

These were either hereditary cholesterol or lifestyle-related cholesterol. The interviewees largely constructed hereditary cholesterol as only being amenable to medication. Nevertheless, there was a high degree of lifestyle talk in their discussions concerning their own responses to having high cholesterol.

Chapter 6 suggested that the prominence of this lifestyle talk, despite the construction of the two distinct categories of cholesterol, reflects the strength of the link between lifestyle and personal responsibility for hypercholesterolaemia in Western culture (Davison et al., 1989, 1991, 1992; Lupton, 1995; Lupton & Chapman, 1995; Petersen & Lupton, 1996; Sachs, 1996). The chapter argued that interviewees were able to establish their own moral probity by oscillating between these different discourses.

Explanations of cases of CHD in people with FH were not necessarily fixed on the hereditary aspect, but drew on factors such as behaviour, physique, stress and temperament. Furthermore, their explanations of other people's CHD did not draw heavily on genetics, suggesting that knowledge of an hereditary basis

to one aspect of CHD does lead people to become more genetic in their outlook in general. These data suggested that the model of the coronary candidate (Davison et al., 1989, 1991, 1992) is very tenacious, even in the face of specific knowledge about hereditary explanations.

FH was situated within the realm of normal, acceptable, unavoidable and treatable illness and on this basis interviewees drew a firm boundary between their condition and other genetic diseases. Reproductive decision-making was not seen as a relevant theme in relation to FH. There was however, much talk about interviewees' actions concerned with taking care of their offspring, both as children and as adults. There was a much looser sense of obligation to inform and encourage action in their wider kin and some interviewees reported adopting the role of cholesterol or FH information-giver with friends, acquaintances and even strangers. As Hallowell (1999) also observed in her study, it is difficult to separate the degree to which responsibilities to others were based specifically on genetic connections or more general obligations based on social connections. Overall, responsibilities concerning *taking care of oneself* were a more prominent feature of the interviewees' talk about FH, than responsibilities in relation to others. This suggests that FH was not predominantly constructed through a genetic lens.

**Summing up - to what extent do FH patients and biomedical professionals construct FH and CHD as genetic conditions?**

Coming back to the overarching question this thesis asks, the research suggests that biomedical constructions of CHD are heterogeneous and that genetic

models represent just one of a number of strands of research in this field. Biomedical professionals involved in HEART UK and patients with experience of FH do not produce genetically-focussed accounts of CHD. Lifestyle continues to provide the dominant discourse. Furthermore, FH is neither understood nor managed within a strong genetic frame.

### **The role played by patients in the construction of discourses about FH and CHD within HEART UK**

The thesis started out by viewing the FHA, the pre-runner to HEART UK, as a potential site of lay knowledge and action and it was selected as a research site on this basis. However, as Chapters 1 and 3 have discussed, the research was reshaped due to the founding of HEART UK, the newly established hybrid organisation that aims to fulfil both the role of patients' association and professional body. One aim of this thesis was, therefore, to establish how the data about HEART UK should be understood, in other words, the degree to which its discourses and activities are indicative of specifically lay constructions. The analysis suggests that lay people were instrumental in founding both the FHA and the Simon Broome Heart Research Trust, and continue to influence the direction and activities of HEART UK. However, Chapter 5 observed that the FHA followed a recognised trajectory from being founded and run by lay people on a voluntary basis, to becoming increasingly professionalised (Rabeharisoa, 2003; Wood, 2000). The data suggest that HEART UK currently has low levels of grass-roots activities, and professional members involved in the management of the organisation far outnumber the lay members. In short, the organisation is somewhat professionally dominated.



The data suggested that lay views may feed into the organisation in quite a loose way, through the collation of queries to the support and information services. However, the organisation could not be said to be influenced by a collective lay discourse in any obvious way.

Interviewees recognised an important role for lay/patient trustees in both contributing their experiential knowledge and their own professional skills, and they contributed particularly to overseeing the work of the patient information and support services and the legal, financial and business aspects of the charity. The data suggest that a division of labour was maintained within the organisation, with the expectation, in the main, that scientific and technical matters would be left to the biomedical professionals. In sum, the data about HEART UK's current activities and discourses should be seen as reflecting professional constructions of FH and CHD as much, if not more than, lay constructions.

## **8.2 PATIENTS' ASSOCIATIONS, KNOWLEDGE AND EXPERTISE**

This section discusses what the findings contribute to discussions about expertise and the emergence of novel forms of knowledge production and dissemination through the work of lay health groups. The analysis suggests that in the early years, the FHA was strongly patient-driven and one of its major aims was to gain recognition of an under-recognised and contested disease category, providing a challenge to contemporaneous medical knowledge and practice. This is not a particularly novel finding in relation to lay health groups. It illustrates one of the main aims of lay health advocacy

groups described by Brown (1995b) and has been discussed in relation to several other conditions (see for example Arksey, 1994; Fox, 1989; Johnson & Hufbauer, 1982; Scott, 1990)

The merge between a patient and professional association at first sight seems like an innovative development with potential to lead to novel forms of knowledge production and collaboration. Drawing on Rabeharisoa & Callon's (Rabeharisoa, 2003; Rabeharisoa & Callon, 2002) model of the partnership organisation, the merge between the patient and professional associations to form HEART UK seemed to provide a potential structure in which collective lay knowledge, collated from patient members' experiences, might interface with biomedical knowledge and in which patients might have some control concerning research policies. The research asked how HEART UK interviewees constructed the role of patients, and how the patients' knowledge is incorporated into the work of the new organisation. The analysis suggests that the organisation recognises a role for patients' experiential knowledge, but at the present time a division of labour is maintained, with lay committee members contributing to decisions about the work of patient services and providing financial and business expertise. Matters concerning biomedical practice and research are delegated to biomedical professionals. The reasons given for the merge were largely instrumental, driven by the potential for increased funds and influence, rather than any specific benefits of collaboration between patients and professionals. The merge can be seen as a political move concerned with gaining increased credibility. Despite the merge, current

relations fit with the idea of the ‘auxilliary’ association described by Rabeharisoa & Callon (Rabeharisoa, 2003; Rabeharisoa & Callon, 2002).

These data suggest that the transformation of expertise that may be emerging in relation to other conditions such as HIV/AIDs, Muscular Dystrophy and PXE (Epstein, 1995; Heath et al., 2004; Rabeharisoa, 2003; Rabeharisoa & Callon, 2002; Rose & Novas, 2004) are not seen in the area of FH. It can be argued that the recognition of patients’ experiential knowledge within the organisation is indicative of the ascendancy of this notion more generally within health policy. This does not appear to challenge the authority of biomedical experts. Matters concerning biomedical research and practice were largely seen as a separate domain. One can argue that the ascendance of the idea of experiential knowledge and the UK government’s involvement agenda, in fact, provided the environment in which such a merge became thinkable. Indeed, interviewees reported that a similar development had occurred in the field of diabetes resulting in the formation of Diabetes UK. It would be instructive to explore the background to this merge and the relationships embedded within Diabetes UK, to compare with the findings of this present study.

Furthermore, the analysis suggests that although HEART UK provides support and information to patients and is seen as representing patients in consultative work, there is currently limited patient input into the running of the organisation. Data suggest that even before the merge, the FHA was highly professionalised, with relatively low levels of grass-roots activities. It was further suggested that in the latter years of the FHA, several of the trustees and

the last two Chairs were biomedical professionals. The current rhetoric within HEART UK of a need for increased patient involvement was not, by their own accounts, being driven by the demands of FH patients. Indeed, interviewees commented on the difficulty of recruiting and retaining patient members. It also did not seem to be driven by any particular commitment to patient expertise, but seemed to be largely driven by the demands of government for patient involvement. Wood's (2000) study of patients' associations highlighted the difficulty of defining such organisations. He talks of basing his inclusion criteria partly on whether organisations appeared to be patient-led and independent. It is, perhaps, time for a reappraisal concerning the meaning of patients' associations. One of the key questions concerns whether they should be defined through their aims and activities or through their membership and management.

Many analysts have noted a rise in the number of lay health groups over the last three decades (see for example Allsop et al., 2004; Epstein, 1995; Kelleher, 1994; Rose & Novas, 2004; Wood, 2000), and this may be interpreted as an indication of changing relationships concerning expertise. The present study contributes to a more detailed analysis concerning the exact nature of the relationships embedded within such organisations. In sum, it can be argued that in the case of the FHA and HEART UK, the organisation was only patient-led under particular conditions at a particular historical moment. The early years were characterised by lack of recognition of the cholesterol hypothesis or conditions, insufficient medical support, lack of treatments and lack of a professional field. By the time of the merge, the cholesterol hypothesis and

biomedical expertise about lipid disorders were largely established, and successful treatments were available. The analysis reinforces the argument that the influence of patients and patients' associations depend on the state of a particular field and on the existing players within it (Rabeharisoa & Callon, 2002; von Gizycki, 1987).

### **8.3 GENETIC RESPONSIBILITY AND BIOSOCIALITY**

The thesis has drawn on the notion of genetic responsibility as one of the ways of analysing whether FH is constructed through a genetic frame. This section turns this analysis around to discuss what this study can say about the notion of genetic responsibility. Although Novas and Rose (2000) reject the idea of geneticisation, they are still committed to the idea that genetic knowledge transforms individuals' subjectivities in some way. They have argued that genetic information places individuals in a web of relations to other people, and discuss this particularly in terms of life plans such as decisions about getting married, having children, careers and finances. They also link the idea of genetic responsibility to wider notions of biological citizenship and biosociality, arguing that people are increasingly joining into groups based on shared biological identities and that activism in relation to such identities can be constructed as an obligation. The case of FH did not seem to fit neatly within Novas and Rose's (Novas & Rose, 2000; Rose, N., 2001; Rose & Novas, 2004) notion of genetic prudence, or active biomedical citizenship.

It is notable that Novas and Rose's (Novas & Rose, 2000; Rose & Novas, 2004) discussions of genetic prudence often enrol the example of Huntington's

disease. This can be characterised by the possible identification of genetic risk in the absence of discernable markers of disease processes or preventative and therapeutic options. Under these circumstances one can imagine that taking a genetic test is concerned almost entirely with the social consequences of being at risk of the condition. In this situation, issues concerning ‘marriage, procreation, financial planning, inheritance and career’ (Rose, N., 2001: 19) may be relevant. FH is different in two major respects. First, in the UK it is largely diagnosed through cholesterol testing. Patients know they are not just potentially at risk; cholesterol testing provides an indication of bodily status that is given on an on-going basis, although this still only signifies a risk of CHD. Second this cholesterol test can lead to immediate therapeutic consequences that significantly decrease the risk of CHD, and cholesterol treatments are not particularly intrusive. It should perhaps not be surprising that FH patients talked a great deal about lifestyle, medication and management of their FH and very little about their ideas about their life plans in the light of the diagnosis or of informing their kin. This suggests that the notion of genetic prudence as defined by Novas and Rose may only be relevant to a small subset of serious genetic diseases where a genetic test is available, but biological markers of risk are absent and preventative or treatment options are lacking. Drawing on the findings of the present study, this notion may be of little relevance when talking of genetic risk of a common and treatable disorders such as CHD, diabetes or asthma.

Rose and Novas (2004) have suggested that biosociality may be specific to certain times and places and Rapp (2001) has commented on the influence of

class and ethnicity/race. Only Allsop et al. (2004) provide any suggestion that biosociality might also be specific to certain biological states or conditions. In the case of FH, trustees of HEART UK may construct membership of this charity as an obligation, but they also reported difficulties in recruiting and retaining patient members. There were also apparently no grassroots activities such as local support groups or web forums in which 'ordinary' patients might talk with each other. These data, and the talk of HEART UK members and 'ordinary' FH patients all suggest that FH is not associated with a strong, specific disease identity, and patients also aligned themselves loosely to a wider 'community' of people with hypercholesterolemia or people with CHD. In short, while interviewees expressed a strong sense of obligation to take care of themselves by adhering to appropriate 'lifestyle' regimes, taking medication and attending medical appointments, the condition did not appear to draw individuals into a 'responsible community of biological citizens' (Rose & Novas, 2004: 451).

It seems that an expectation of active biological citizenship and biosociality are embedded within UK health policy relating to patient involvement and was also rehearsed by members of HEART UK. It was not, however, a dominant part of 'ordinary' patients' constructions of their condition. This can again be seen as related to the characteristics of FH i.e. it is treatable and relatively unobtrusive, and perhaps also to the 'ordinariness' of hypercholesterolemia and CHD. This is just one case study and it is possible that FH is atypical, but the findings here suggest that Rose and Novas may overstate the significance of

biosociality. Future analysis might help to characterise the kinds of conditions where it is most and least evident.

#### **8.4 WHAT CAN BE SAID ABOUT THE GENETICISATION THESIS?**

The findings of this study challenge the geneticisation thesis in a number of important respects. Chapter 3 laid out a set of key claims incorporated within the idea of geneticisation. These were drawn on in each chapter in order to help to structure the analysis. This section will draw together the arguments relating to each of these claims:

**Genetics is the dominant discourse:** The analysis suggests that there is a range of models of CHD in the biomedical literature and that genetic models are not dominant in the research of HEART UK members, in the talk or official texts of HEART UK, or in the talk of FH patients. Indeed, the influence of genetic variations on CHD risks in general was practically absent from all these discourses. HEART UK continued to emphasise the established risk factors for CHD, framed mainly as ‘lifestyle’ factors, while FH patients continued to draw on established lay models of CHD embodied by the idea of the coronary candidate. Furthermore, neither the Simon Broome publications, HEART UK, nor FH patients constructed FH through a strongly genetic frame and there did not seem to be a specific FH identity. The activities of the FHA and HEART UK could be said to have become less rather more focussed on genetics over the last two decades, as the organisation broadened its remit to include cholesterol and CVD risks in general. This is the opposite of geneticisation.



**Genetic discourses are reductionist and deterministic:** Neither biomedical nor lay constructions framed genes as necessarily the determining factor in cases of CHD for people with FH. This was demonstrated in the Simon Broome Group's interest in explaining differential rates of CHD in FH as well as patients' accounts of their own and their relatives' CHD.

**Society is becoming stratified along genetic lines:** Both HEART UK and 'ordinary' patients with FH constructed an aetiological distinction between hereditary and 'lifestyle-induced' hypercholesterolaemia. This conceptual distinction enabled people with FH to construct themselves as less accountable for their condition than other people. However, distinctions on genetic grounds were not necessarily constructed as useful in practical terms. While the Simon Broome publications have discussed the possibility of incorporating DNA-based information into the diagnostic criteria for FH, current diagnostic criteria in the UK remain largely based on clinical indicators. HEART UK interviewees were divided about the benefits of distinguishing between patients with raised cholesterol on the basis of their DNA and often privileged clinical status. Divisions on the basis of genetics were not seen in the activities of HEART UK, which provides information and support to all people with raised cholesterol regardless of the aetiology. Patients with FH also did not necessarily draw a sharp line between their own raised cholesterol and other people's. Some described talking with other people to raise awareness and encourage action in relation to cholesterol in general.

**Geneticisation affects concepts of normality and abnormality:** Patients with FH largely rejected the genetic framing of the condition in terms of reproductive decisions, drawing a line between it and ‘serious’ genetic disease. They constructed FH as part of normal, acceptable and unavoidable illness and this was strongly related to the treatability and manageability of the condition. This supports Lambert and Rose’s (1996: 79) observation that the hereditary lipid disorders are seen as part of the normal spectrum of human health and illness; they are part of ‘the normality of human imperfection’. Furthermore, the idea of reproductive decision making in regard of FH was almost entirely absent from the biomedical literature and the talk of HEART UK interviewees.

**Geneticisation privatises and individualises health risks and responsibility, focussing attention on biological rather than social conditions associated with illness:** It is true that models of CHD constructed through the research of HEART UK members, the Simon Broome publications and HEART UK’s texts and activities tended to individualise responsibility for CHD risks, paying little attention to the structural, social and cultural context of these risks. In this case, however, this has little to do with a rise of genetic discourses about CHD, as such discourses were not evident in the sites researched. The findings reinforce the argument made in Chapter 3 that the geneticisation thesis is part of a wider critique concerning the appropriate way to define and manage health problems. Abby Lippman, like other social scientists, public health physicians and epidemiologists, is trying to draw attention to the social and structural factors that contribute to disease and is critical of preventative strategies that focus on individual action and responsibility. The data show that the same

criticism Lippman makes of geneticists concerning the locus of responsibility for preventative action, is also levelled by epidemiologists and public health physicians (Beaglehole & Magnus, 2002b; Marmot, 2002) at cardiologists.

**Geneticisation leads to the increased use of genetic technologies in health**

**care:** As already discussed, in the UK, FH is diagnosed largely on the basis of clinical indicators, i.e. raised cholesterol, family history, and physical signs.

DNA-testing has not been widely introduced in the UK. The discussions of the Simon Brome register Group and HEART UK interviewees suggest that even if the use of DNA diagnostics were to widen, they would be unlikely to replace clinical methods of diagnosing FH. Furthermore, genetic technologies have, so far, had little impact on the diagnosis and treatment of CHD more widely.

**Genetic discourses suggest that genetic research is imperative for future**

**health improvements:** The review of the biomedical literature suggests that there is a range of research interests concerning CHD. The idea that genetic research is imperative was found in the review paper written by geneticists, as might be expected, but was absent elsewhere. Research by HEART UK members was rarely concerned with genetic variations in CHD, but was much more concerned with the molecular pathways involved in atherosclerosis and the management and treatment of lipid disorders. One recurrent theme in relation to both CHD and FH was that health improvements could accrue through better application of current knowledge and practice. This was also manifest in HEART UK's focus on 'lifestyle' aspects of CHD and the better

identification and management of FH based on current recommendations. In the sites studied there was no drive for genetic research.

**Geneticisation reflects the power of geneticists to identify and classify**

**health problems:** This analysis has argued that there are a number of disciplines with an interest in CHD including geneticists, epidemiologists, public health physicians, cardiologists and metabolic specialists. This is probably not an exhaustive list. It has suggested that it may be fruitful to consider the notion of geneticisation as part of a boundary dispute between epidemiologists and other disciplines, and also that there may be a number of boundary disputes concerned with constructions of CHD and FH. The data have shown that epidemiologists and public health physicians including Lippman, Beaglehole, Magnus and Marmot privilege socio-structural understandings of CHD risk and are critical of both geneticists and clinicians for individualising CHD prevention. It has also suggested that there may be disputes between metabolic specialists and geneticists over the definition and diagnosis of FH as well as historical disputes between metabolic specialists and cardiologists over the acceptance of the cholesterol hypothesis. These disputes are all concerned with who has the jurisdiction to decide what kinds of conditions CHD and FH are, and the best ways of managing them. While the analysis has suggested that biomedical discourses about the aetiology and management of CHD and FH are heterogeneous, it has also recognised that these discourses do not necessarily divide strictly along disciplinary lines. The data illustrated, for example, that some clinicians are in favour of DNA diagnosis for FH. It must, in any case, be recognised that geneticists are just

one of a number of important players in the field of CHD.

Differences in disciplinary perspectives concerning CHD should, in some senses, come as no surprise. These reflect the different concerns, objectives and professional practices of the different disciplines. While molecular geneticists are concerned with processes at the molecular level, epidemiologists are largely concerned with disease prevention at the population level and clinicians are concerned with practical interventions at the individual level. Arguments in this thesis are premised on the idea that geneticisation is not just represented through discourse, but it is embodied through medical practices and technologies. Kerr (2000) and Shaw (2003) have demonstrated that even for so called established genetic conditions such as cystic fibrosis and the dysmorphologies, genetic information is not necessarily of utility to clinicians. Hedgecoe (2002, 2004b) has illustrated that there were disciplinary differences between scientists and clinicians concerning diagnostic categories for diabetes and has shown that clinicians were resistant to the inclusion of genetic testing in the diagnosis of Alzheimer's disease. The current research has demonstrated that in the case of FH, as well, clinicians were ambivalent about the utility of genetic information in the diagnosis of the condition. In more general terms, Cox & Starzomski (2003) and Hall (2004) have suggested that clinicians may be reluctant to focus on genetic elements when discussing common diseases with their patients. This may be, in part, because it currently makes little difference to the therapeutic treatments they can offer. The same seems to be true in the area of FH, where the focus was on treatments and risk factors seen as 'modifiable'.

### **Why is geneticisation not evident in this case?**

Drawing these different areas together, the analysis suggests that the expectations embodied by the geneticisation thesis do not so far appear to have been realised in the area of CHD and FH. There may be a number of factors that contribute to explaining why genetic constructions of FH are not prominent in this study and why there was not a strong sense of an FH identity. These relate to the particular characteristics of the condition, the setting where health care is provided and the way FH and CHD are understood. First FH is treatable and, of itself, has no symptoms. It was perceived by patients to be highly manageable and not serious enough to be classified in the same category as other genetic conditions. Second, the clinical aspects of FH are not distinct. It is monitored, treated and managed in much the same way as 'polygenic' hypercholesterolaemia. Third, the condition is treated by metabolic specialists rather than through the genetic services, drawing on the techniques and practices of metabolic medicine rather than of clinical genetics. Fourth, there are already well-established biomedical and lay models of CHD that focus on 'lifestyle' or 'modifiable' factors. Lay models already included a general sense that CHD can run in families, and the diagnosis of FH seemed often to be understood in this general way, rather than as a specific Mendelian condition with predictable transmission patterns. Fifth, prior to diagnosis, patients with FH had not necessarily picked up on a family history of early CHD. This was perhaps due to the commonness of CHD in general and the availability of other explanations.

Cox and Starzomski (2003) enumerate a similar list of factors to explain the lack of geneticisation in the case of PKD, suggesting additionally that the lack of a specific disease support group may contribute to the lack of a genetic construction. The analysis of HEART UK showed that one cannot assume that diseases will be constructed in a genetic way, even where there is a specific patients' association. These associations must be seen as sites where disease constructions are negotiated, subject to the same influences as other sites. Hedgecoe, (2002: 9) talks of 'geneticisation by stealth' in the case of diabetes, suggests that diabetes is constructed in some quarters as a paradigmatic multifactorial genetic disease, and demonstrates how its classification came to have a genetic basis. Nevertheless, there is a strong thread of emphasis on 'lifestyle' factors in explaining Type 2 diabetes and links to the rhetoric of an 'obesity epidemic'. There are many parallels between constructions of diabetes and CHD and it seems likely that the same heterogeneity of discourses and practices will be evident.

In sum, this thesis has argued that geneticisation is not evident in the case of FH and that factors such as the availability of effective therapeutics, the sites where care takes place, the disciplines and technologies involved and the existing lay models of disease may have important implications for the construction of a particular field. If geneticisation is evident in neither the case of FH nor PKD, where there are established hereditary links to CHD and kidney disease, this raises questions about the impact of genetics in relation to other common complex conditions, the utility of the concept and the kinds of phenomena to which it can be applied.

## **8.5 IMPLICATIONS FOR POLICY AND PRACTICE.**

This section relates the findings of the study, in the broadest terms, to health policy concerning genetics, making some fairly general observations. It then discusses the findings in relation to two very specific aspects of practice concerning FH.

Policy and initiatives in the UK relating to the development and use of genetic knowledge and technologies within healthcare embody high hopes and expectations for this knowledge. In the forward to the genetics White Paper (Cm 5791 - II, 2003: 5), John Reid, the former Secretary of State for Health, talks of both ‘the promise of more personalised healthcare with prevention and treatment tailored according to a person’s individual genetic profile’ and of the likelihood that genetic based healthcare will ‘expand and permeate every area of medicine as new genetic tests and therapies come on stream’. These expectations embody a set of assumptions about the way biomedical professionals and lay people will be likely to view this knowledge. They assume both that biomedical professionals in ‘every area of medicine’ will welcome genetic knowledge and technologies and that lay people will seek personalised preventative strategies. The thesis provides a number of insights on these areas that may be useful to policy makers involved with genetics and healthcare.

First, as the previous discussion has highlighted, professional constructions of disease are complex and heterogeneous. They relate to specific practices and



cultures of particular biomedical fields. Different professionals are likely to have different priorities. The analysis has suggested that, for example, DNA-based testing will not be universally welcomed by clinicians. In the case of FH, although DNA-based testing was seen as helpful by some of the clinicians, it was certainly not seen as definitive and would be unlikely to replace clinical criteria. In some cases clinicians prioritise clinical criteria, partly because of the uncertainties concerning the relationship between identifying a mutation and the clinical expression of the condition. Other studies have also demonstrated that clinicians may question the utility of genetic testing, even for so called classic genetic conditions (Hedgecoe, 2004b; Kerr, 2000; Shaw, 2003). Policy makers should be aware of this complexity. Lack of clinical demand would not necessarily be indicative of lack of knowledge about genetics, but would be likely to reflect different priorities and aims of the clinicians involved<sup>18</sup>. Findings of this and other studies, therefore, suggest that policy makers may need to develop more realistic expectations about the utility of genetic information and technologies in clinical practice.

Second, the vision of a personalised preventative strategy rests on the expectation that lay people will want and choose to engage with such information and adhere to recommended strategies. Marteau & Lerman (2001) suggested that there was a paucity of data concerning people's actual rather than intended behaviours following the provision of genetic risk information. This thesis suggests that in the case of FH, the patients who are lipid clinic

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<sup>18</sup> It should be recognised that even among geneticists, doubts have been raised about the predictive value and clinical utility of genetic testing for common complex diseases, particularly where multiple genes are involved. See, for example, Holtzman & Marteau, (2000), Janssens et al. (2004), Wilkie (2001).

attendees had been largely unfazed by their diagnosis and that they had adopted the preventative stance embodied within policy expectations. The analysis has suggested that interviewees' construction of FH as largely unproblematic was tied up with the availability of effective medications; they saw FH as highly treatable. This research cannot predict the uptake of genetic or other health related risk information where prevention is based entirely on the adoption of 'lifestyle' interventions in the absence of specific medications or other medical interventions.

Furthermore, although interviewees presented having FH as largely unproblematic, their talk about their relatives and their own younger lives sometimes suggested that people may resist genetic or other health risk information. This was often related to age and lifecourse. Interviewees suggested that young people, in particular, may be less willing to engage with such information, because 'when you're young you think you'll live for ever'. Lawton (2002) has suggested that the degree to which people are conscious of health and the possibility of ill-health is related to age and, more importantly the embodied experience of ill-health. This may relate to people's receptiveness to predictive health information. Policy makers should be aware that people are unlikely to be universally receptive to the idea of genetic susceptibility testing and that this may depend on the availability of therapeutics or to demographic or other characteristics of the patient.

This section will now turn to two very specific points concerning FH arising out of the data. As has already been argued, this research suggested people

who attend lipid clinics do not find the management of their condition problematic. Nevertheless, a small number of people who reported that they were diagnosed in childhood or early adulthood reflected that they had perceived this as an unwanted intervention at the time, and their discussion suggested perhaps a need for a certain amount of support for children. Diagnosis and care of children was certainly one of the more prominent areas of interviewees' discussions about living with FH. Although there is guidance concerning the diagnosis and management of FH in children, it appears to make little reference to any psychological aspects of this. The Department of Health's Cascade Screening Project does not discuss whether children will be included or any special issues that may arise from this (Hadfield & Humphries, 2004). National guidance concerning genetic testing of children for adult onset conditions (Advisory Committee on Genetic Testing, 1998) does not specifically discuss FH, but provides no obstacle to its early screening, precluding only the testing of young children for conditions where therapies are not available. Policy makers and practitioners may wish to reflect on services and guidelines concerning the diagnosis and treatment of children. There may be an argument for providing support to families, particularly in cases where the parents have been previously unaware of a family history of CHD or hypercholesterolemia. It is notable that HEART UK has already recognised this area and has been involved with running two children's lipid clinic projects.

The second point arising from the study relates to a fairly minor observation in the methods chapter. This noted that within the sample of 31 lipid clinic

patients, there was a marked difference in experience of CHD by occupational category, with those with managerial, professional and intermediate occupations having less CHD than those with manual and routine occupations. Comments on this observation are necessarily very speculative because of the small sample size. If these patterns were replicated within the whole lipid clinic population this might suggest that people with FH who work in manual and routine occupations have higher rates of CHD or perhaps that they enter the clinic at later stages of their FH career. Chapter 3 noted that there does not appear to have been any analyses by occupation of rates of CHD in people with FH. Should this pattern be seen across the whole clinic population, this would be an argument for further analysis to explore whether this was related to access to service and/or differences in morbidity and mortality. In either case, the issue of occupational class and FH would seem to warrant some further analysis.

## **8.6 QUESTIONS RAISED AND FUTURE RESEARCH**

The analysis raises a number of additional questions that point to potential areas of research that would build on the thesis.

### **Biomedical research on CHD**

The thesis provided an analysis of the rhetoric of a small number of commentary papers about CHD and an overview of the research of one very specific group of CHD researchers. There is scope for a wider analysis of research and research funding in the area of CHD. It would be fruitful to adopt both quantitative and qualitative methods in this area.

### **Historical context of genetic models of CHD and hypercholesterolaemia**

The analysis of biomedical constructions of CHD undertaken in this thesis focussed on a limited, recent time period and it is not clear, for example, when or how the notion of ‘polygenic’ hypercholesterolaemia or a polygenic model of CHD emerged. There is scope for a more detailed socio-historical work in this area to show how and why a genetic model of CHD came about and how this relates to the emergence of the cholesterol hypothesis and other bodies of research in CHD.

### **The clinical consultation as a site where FH is constructed.**

The thesis focussed mainly on the biomedical literature and FH patients’ accounts of living with FH, and included the talk of a small number of clinicians about their clinical practice. It has not looked at how clinicians and patients construct FH through clinical consultations. It would be possible to extend this research to undertake an observational study, with the aims of analysing how FH is constructed during these consultations and how this relates to patients’ accounts of FH provided in this thesis.

### **Patients’ constructions of ‘polygenic’ hypercholesterolaemia**

The thesis has drawn on the work of Davison and his colleagues (Davison et al., 1989, 1991, 1992) on lay constructions of CHD to provide comparison data. Their analysis was based on a study of the general population. It could be argued that more direct comparison data would be provided through a study of how patients with ‘polygenic’ hypercholesterolaemia account for their

condition. This might also help to clarify whether responsibility to others, such as encouraging kin and other people to be tested, relates directly to a genetic connection or to a social connection.

### **International comparisons**

It has already been noted that practice concerning FH may vary trans-nationally. The Netherlands stands in particular contrast to the UK because it has implemented a national policy of cascade screening, based on genetic diagnosis which is undertaken through the genetic services. It would be instructive to analyse the context and circumstances through which this policy came about and the similarities and difference to the UK situation. Interviews with FH patients in the Netherlands would provide comparison data that could provide evidence about the possible influence of screening and DNA testing to patients' constructions of FH and responsibility.

## **8.7 FINAL REFLECTIONS**

Since its introduction by Lippman in the early 1990s, the concept of geneticisation has been widely adopted across a number of disciplines. It has come to hold a range of meanings from the increasing prominence of genetic explanations in the media, professional or public discourses, to a full-scale critique of the implications of genetic ways of thinking and doing. Chapter 3 argued that many of the social analysts who have enrolled or discussed geneticisation, both its supporters and detractors, have assumed, without questioning, that genetic discourses and practices are spreading. In doing so, they have contributed to a set of expectations about the great influence that

genetics and geneticists have or will have in health care and beyond. While some of these analysts see this influence as negative and others less so, they have all reinforced the image of strong 'genetic agency' (Webster, 2005). This image is not supported by this study of patient and professional constructions of FH and CHD. The thesis contributes to a more nuanced understanding of the place of genetics in healthcare and in society.

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## APPENDIX 1: RECENT PAPERS ON GENETICISATION OR CITING LIPPMAN 1991/1992

### A: PAPERS ON GENETICISATION 2002 –2004, IN ENGLISH<sup>19</sup>

paper	Area or illness	Levels of data	site	focus	Argument
Ellison & Jones (2002)	Social identities	Discussion piece		classification of social identity (race/ethnicity) in genetic research and testing.	'a <i>collective</i> geneticization of social identities shifts responsibility for social inequalities in health on shared values, beliefs and behaviours' (p.267 original emphasis)
Gibbon (2002) <sup>20</sup>	Breast cancer	Patient & clinical discourse	Clinical genetics	use of family trees in clinical practice	Critique of the 'impact' approach of social studies about genetics. Patients had an active role and investment in the process. There is a need to 'rethink normative notions of geneticization and medicalization' (p. 454 )
Hedgecoe (2002)	Diabetes	Scientific discourse	review papers	Classification of diabetes	Genetic information was pivotal to the reclassification of diabetes on aetiological rather than clinical grounds. Demonstrates 'geneticization by stealth' where genetic explanations enter into classification at early stage, only becoming apparent later.
Hedgecoe (2003a).	Cystic fibrosis	Scientific discourse	review papers	Classification of CF	CBAVD, a form of male infertility, has been redefined as part of a CF-continuum, leading to tensions between this new categorisation and clinical diagnostic practices.

<sup>19</sup> Identified through search of 'geneticisation' or 'geneticization' of WOS and ASSIA, undertaken on 19/4/05

<sup>20</sup> Identified through Google web search on 1/3/05, not through WOS/ASSIA search

<b>paper</b>	<b>Area or illness</b>	<b>Levels of data</b>	<b>site</b>	<b>focus</b>	<b>Argument</b>
Shaw (2003).	dysmorphology	Clinical practice	Clinical genetics.	Diagnostic process in clinical genetics	Clinical geneticists do not prioritise genotypic information over phenotypic in diagnosis. As in other diagnostic settings, genetic diagnosis is a clinical process, not a form of genetic reductionism.
ten Have (2003)		Discussion piece		Bioethical discussion of geneticisation	Geneticisation is useful as a tool to refocus bioethical analysis and can be analysed on a number of levels.
Wilcox (2003)	sexuality	Media discourses	TV & print media	Media coverage of biological research on sexuality	There are multiple meanings attached to biology, genetics and being 'born with' a characteristic. These should not necessarily be read as an indication of geneticization.
Chadwick & Aindow (2004)	Psychiatric illness	Discussion piece		treatment and research ethics in psychiatric illness	Discusses geneticization because it has 'far-reaching implications because of the potential for predictive testing and gene therapy' p293.
Cox & Starzomski (2003)	Polycystic Kidney Disease	Patient & clinical discourses	nephrology & clinical genetics	the social construction and clinical management of PKD	'recent advances in genetic knowledge and techniques [have] had a minimal impact on the clinical management and social construction of PKD' (p161). Discusses factors that may mitigate geneticization.
Hall (2004)	Heart disease	Patient & clinical discourses	coronary care unit	the 'seeming geneticization' of heart disease	The process of geneticization is not straightforward as imagined by Lippman. In the clinical setting, professionals foreground lifestyle, because of concern about patient fatalism, whereas patients would have liked an acknowledgement of a genetic contribution.
Hedgecoe (2004a)		Discussion piece		Reply to Kerr (2004)	Contests methodological questions and substance of argument.

<b>paper</b>	<b>Area or illness</b>	<b>Levels of data</b>	<b>site</b>	<b>focus</b>	<b>Argument</b>
Kerr (2004)		Discussion piece		Critique of Hedgecoe (2003)	Hedgecoe's analysis overstates the impact of molecular knowledge on the definition and diagnosis of CF and he produces an ahistorical and inflexible model.
Melendro-Oliver (2004)	Common acquired & complex behavioural traits	Media & scientific discourse	journalistic & scientific texts.	The redefinition of genetic disease	The Human Genome Project challenges reductionist and deterministic models of disease and behaviours, the shift to genomics has not seen an end of determinism.
Raz & Atar (2004).	Spouse selection & prenatal screening	Lay /patient discourses	Community genetic services	Introduction of community genetics to Bedouin Arabs in Israel.	This community were not passive, but actively participated in response to this new service, with some accepting it, some rejecting it and some using it with a different rationale (for their own ends)

**B: ADDITIONAL PAPERS IN 2004 CITING LIPPMAN (1991) OR LIPPMAN (1992)<sup>21</sup>**

<b>Paper</b>	<b>Main focus</b>	<b>Main use of Lippman 1991/1992</b>	<b>Critiques Lippman?</b>	<b>Geneticisation cited?</b>
Baylis & Robert (2004)	Ethical analysis of genetic enhancements	<ul style="list-style-type: none"> <li>• Diversion of resources</li> <li>• Individualisation of responsibility</li> </ul>	No	No
Ceccarelli (2004)	Analysis of metaphors used in speeches at completion of HGP	<ul style="list-style-type: none"> <li>• metaphors &amp; reductionism/determinism</li> </ul>	Yes	No
Condit (2004)	Review of research on media and lay discourses about genetics	<ul style="list-style-type: none"> <li>• metaphors &amp; reductionism/determinism</li> </ul>	Yes	No
Greco (2004)	Analysis of the indeterminacy of health as a political issue.	<ul style="list-style-type: none"> <li>• Scientific discourse concerning genetics</li> <li>• Normality/abnormality</li> </ul>	Yes	No
Helén (2004)	Analysis of new reproductive technologies, diagnosis and abortion from a governmentality perspective	<ul style="list-style-type: none"> <li>• Focus of high-tech medicine on prediction and prevention</li> <li>• Agency of pregnant women</li> </ul>	Yes	Yes
Kirby (2004)	Analysis of the film 'GATTACA' and its implications for race and genetics	<ul style="list-style-type: none"> <li>• Societal trend toward genetic reductionism.</li> </ul>	No	Yes
Koch (2004)	The different meanings of eugenics and its use by both critic and supporters of new genetic technologies	<ul style="list-style-type: none"> <li>• genetics as reductionist and undesirable</li> </ul>	Yes	No
Lemke (2004)	Analysis of genetic risks from a governmentality perspective.	<ul style="list-style-type: none"> <li>• 'Geneticization of society' (p550)</li> <li>• Genetic reductionism in scientific discourses</li> </ul>	Yes	Yes

<sup>21</sup> Identified using WOS on 19/04/05

<b>Paper</b>	<b>Main focus</b>	<b>Main use of Lippman 1991/1992</b>	<b>Critiques Lippman?</b>	<b>Geneticisation cited?</b>
Robert & Smith (2004)	Ethical analysis of the Environmental Genome Project	<ul style="list-style-type: none"> <li>• Analysis of complex diseases, not just classic genetic diseases</li> <li>• Individualisation of risk and victim blaming</li> </ul>	No	No
Santos & Maio, (2004)	Analysis of arguments about national identity ensuing after report on a study into the 'genetic origins of Brazilians'	<ul style="list-style-type: none"> <li>• Geneticization as an 'array of transformations and generation of new meanings' resulting from new genetics (p348)</li> </ul>	No	Yes
Schubert (2004)	Ethical analysis of pharmacogenetics	<ul style="list-style-type: none"> <li>• reductionism and determinism in other areas of testing</li> <li>• (pharmaco) genetic identities – new stratification</li> </ul>	No	Yes
Sherwin (2004)	Ethical issues associated with BRCA testing and the implications of susceptibility testing.	<ul style="list-style-type: none"> <li>• Growing 'public sentiment' that all health risks reside in genes.</li> <li>• Reductionism and determinism</li> <li>• Public belief that health improvements will follow directly from genetic knowledge</li> <li>• Diversion of support from other factors</li> </ul>	No	Yes
Shields et al. (2004)	Ethical and social implications of research into the genetics of smoking	<ul style="list-style-type: none"> <li>• Role of media in constructing promise of genetics</li> <li>• 'overly geneticized view of human identity, behaviour and health' (p684)</li> <li>• geneticization as a problem of public understanding of science</li> </ul>	No	Yes

<b>Paper</b>	<b>Main focus</b>	<b>Main use of Lippman 1991/1992</b>	<b>Critiques Lippman?</b>	<b>Geneticisation cited?</b>
Surbone (2004)	Moral implications of BRCA testing	<ul style="list-style-type: none"> <li>the privileging of the role of genes in disease causation, in both medical practice and social attitudes.</li> </ul>	No	Yes
van Delden et al. (2004)	Ethical implications of pharmacogenetics	<ul style="list-style-type: none"> <li>uptake of genetic determinism by both patients and healthcare professionals</li> <li>both increasing and decreasing individual responsibility</li> </ul>	No	Yes

## **APPENDIX 2: RECRUITMENT DOCUMENTS FOR HEART UK INTERVIEWS**

### **A: LETTER OF INVITATION FOR HEART UK INTERVIEWEES**

Dear [name]

#### **The Role of HEART UK**

I am writing to you at the suggestion of Julie Foxton to see if you might be able to help me with my research. This is concerned with the work of patients' associations in supporting patients with familial hypercholesterolaemia. I am particularly interested in the work of HEART UK. The research is funded by the Economic and Social Research Council and has been approved by the administration at HEART UK. I have enclosed an information sheet which provides further details.

I am hoping to undertake a limited number of interviews with professionals and patients who are involved in the activities of HEART UK, with the aim of finding out more about the work undertaken by the Trust, the policies supported and roles of the different groups within the Trust.

If possible, I would like to meet with you to talk about your work with HEART UK and your involvement in the [name of committee/s]. I estimate that an interview would take an hour at the very most and anything discussed would be in confidence.

I hope you might have time for a brief meeting of this sort. I intend to ring you in the near future to see if this will be possible. Please do not hesitate to contact me if you would like to discuss the research further or you have any queries

Many thanks  
Yours sincerely

Kate Weiner

## **B: INFORMATION SHEET FOR HEART UK INTERVIEWS**

### **How do patients understand familial hypercholesterolaemia? The role of HEART UK**

#### **Background**

Patient associations play an important role in providing information both to patients and health care practitioners and influencing policy and research agendas. They also provide a forum for patients' concerns and a point of contact between patients and clinicians. The valuable work carried out by patient groups is increasingly recognised within government and policy circles. In the light of the merger with the BHA, HEART UK is particularly interesting in the way patients, health professionals and scientists work together within the organisation. Although the Government recently endorsed cascade screening for FH, there has been relatively little research on patients' views on and experiences of FH or other treatable inherited conditions. This study aims to inform health policy and practice concerning the application and extension of susceptibility testing for CHD and shed light on the contribution of patient groups in informing and supporting patients and shaping developments in this area.

#### **The Research**

The research is concerned with the work of HEART UK, the policies and research supported and the role of the different groups within the Trust. It aims, broadly, to answer the following questions

- How and to whom does HEART UK provide support and information?
- What role does the Trust have in shaping policies and research in the areas of CHD and genetics?
- Who contributes to the activities of HEART UK and in what ways?

The research will be based on observations of the main activities of the Trust and on interviews with staff, with professional and patient members involved in the committees, and with a small number of other active patient members. Interviews will address people's work and involvement within the organisation and their reasons for this involvement, the aims and activities of the organisation, the research pursued and supported, the collaborative and advisory work undertaken, the recent history of HEART UK, and hopes and plans for the future.

This research forms part of a wider study concerned with patients' understandings and experiences of FH. In addition to the research outlined above, a number of interviews with patients with FH will be undertaken. The patients will be recruited through a lipid clinic. The interviews will focus on patients' views, concerns and experiences of FH.

#### **The researcher**

This research is being undertaken by Kate Weiner for her PhD and is funded by a research studentship from the Economic and Social Research Council. Kate has several years experience as a research associate in the social policy field. Her first degree was in the biological sciences.

For further details about the research contact:

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## **APPENDIX 3: INTERVIEW GUIDE FOR HEART UK**

### ***About yourself***

- What is your role within HEART UK and what does this involve?
- How did you become involved?

### ***About the organisation***

- What do you see as the main aims of the organisation?
- What are the main activities of the organisation?
- What research has HEART UK been involved in?

### ***Work with other organisations***

- Have you done any advisory or collaborative work on behalf of HEART UK? (With which other organisations, member of any umbrella groups, types of consultations)
- What public campaigns has HEART UK instigated or been involved in?

### ***The past***

- Before the merge, were you involved with the FHA or BHA? (in what ways?)
- Can you tell me anything about the history of HEART UK/FHA/BHA/Simon Broome Heart Research Trust? (origins, aims and activities, people and organisation, links between the organisations, developments)

### ***The merge***

- In your view, how did the merge between FHA and BHA come about?
- What do you think have been the main changes since the merge? (Benefits and disadvantages?)
- In your view, what role do patients play in the organisation? [FOR STAFF AND PROFESSIONALS] How do you see your role in relation to the clinicians and staff? [FOR LAY MEMBERS] (Is it important to have patients on the committees? Why? What do patients bring to the organisation?)

### ***The future***

- What do you see as the main priorities for people with FH?
- What developments would you like to see take place in HEART UK in coming years?
- What research or policies would you like to see HEART UK support or undertake?
- What do you think about DNA diagnostics for FH? – do you see any advantages?

## **APPENDIX 4: RECRUITMENT DOCUMENTS FOR INTERVIEWS WITH LIPID CLINIC PATIENTS**

### **A: LETTER OF INVITATION TO LIPID CLINIC PATIENTS**

Dear

I am writing to invite you to participate in a study that will be taking place in the clinic between January and June 2004. This is concerned with patients' views about and experiences of familial hypercholesterolaemia. Participation will involve being interviewed on a single occasion and will not involve any change to your current care or medication. Please read the enclosed participant information sheet for details of the study and how you were selected.

The study will be undertaken by a PhD student, Kate Weiner. The research has been approved by the [research ethics committee].

I would be grateful if you could complete and return the reply slip in the envelope provided, indicating whether you are willing to participate. Two options are provided for people who are willing to participate. You are asked to choose whether you would prefer to be interviewed at the clinic on the day of your next appointment, or somewhere else, like your workplace or home. If you indicate you are interested in participating, your contact details will be passed on to Ms Weiner. She will contact you regarding the arrangements for the interview. If you do not wish to participate, no further contact regarding this matter will be made.

Thank you in advance

Yours sincerely

[clinician]

---

### **REPLY SLIP**

Participant identification number:

Please tick as appropriate

Yes, I am interested in participating. I would like to be interviewed at the clinic when I come for my next appointment

Yes, I am interested in participating. I would like to be interviewed at a time and place convenient to myself

No, I do not wish to participate in this research.

## **B: PI SHEET FOR LIPID CLINIC PATIENTS**

### **PARTICIPANT INFORMATION SHEET**

#### **Study title: How do patients understand familial hypercholesterolaemia?**

You are being invited to take part in a research study. Before you decide it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with others if you wish. Feel free to contact me if there is anything that is not clear or if you would like more information (see contact details at end of sheet).

Thank you for reading this.

#### **What is the purpose of the study?**

It is possible that in the future genetic susceptibility tests will be developed for complex conditions like heart disease. These would identify people who are at increased risk of illness, but who are currently well. Familial hypercholesterolaemia (FH) provides an example of a relatively common inherited condition which puts people at increased risk of heart disease. There has been remarkably little research on the social issues raised by screening and diagnosis of familial hypercholesterolaemia or other treatable inherited conditions that lead to increased risk of illness.

This research will focus on patients' perspectives of FH and aims to explore how patients understand the condition. It will focus on people's day-to-day experiences of the condition, covering such issues as how patients explain FH in the different situations in which they find themselves and how they see their health status. The study will take place between January and June 2004

#### **Why have I been chosen?**

The research will draw on patients who attend the lipid clinic at [hospital]. Your name was selected randomly from a list of all adults with familial hypercholesterolaemia who are likely to attend the clinic between January and June 2004. You are one of about 30 people in total who have been invited to participate.

#### **Do I have to take part?**

NO. It is up to you whether or not you take part. If you do decide to take part you will be given this information sheet to keep and be asked to sign a consent form. If you decide to take part you are still free to withdraw at any time. If you decide not to take part, or decide to withdraw at any time, you do not have to give a reason and it will not affect the standard of care you receive in any way.

**What will I be asked to do if I take part?**

If you agree to participate, you will be interviewed about your experiences of and views on FH. The interview will be quite informal and conversational in style. It will probably last no longer than one hour. You can choose whether you would prefer to be interviewed at the clinic on the day of your next appointment or somewhere else, like your workplace or home. If you choose to be interviewed at the clinic, I will arrange the interview to take place immediately after your appointment and I will contact you to confirm the arrangements. If you choose to be interviewed elsewhere, I will contact you to arrange where and when to see you. At the interview, I will ask for your permission to tape-record the interview. You can change your mind at any time and the tape will be turned off or destroyed at your request.

**What are the possible benefits of taking part?**

There will be no direct benefit to your care if you take part, but the findings will be helpful to policy makers and health care practitioners who are involved in the development and use of genetic susceptibility testing and screening.

**Will my taking part in this study be kept confidential?**

All information which is collected about you during the course of the research will be kept strictly confidential. The tape recordings of the interviews will be kept in a locked drawer to which only I have access. When I write reports on the study I will not use your name or any information which would identify you. I may use extracts of what you say at the interview, but this will not include anything that might identify you.

**Who is organising and funding the research?**

This research is being undertaken by myself, Kate Weiner, as part of my studies for a PhD in medical sociology. I am funded by a grant from the Economic and Social Research Council. I am a student at Nottingham University, but I work in [city].

**What will happen to the results of the research study?**

The main report from this study will be my PhD.

**Further Information**

If you have any questions about the research or require any further information please feel free to contact me. Please note, I am not a health professional and so I cannot answer any medical questions.

Thanks once again for taking the time to read this.

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**C: CONSENT FORM FOR LIPID CLINIC PATIENTS**

Participant Identification Number:

**CONSENT FORM**

**Study Title: How do patients understand familial hypercholesterolaemia**

**Name of Researcher: Kate Weiner**

1. I confirm that I have read and understand the information sheet dated August 2003 (version 1) for the above study and have had the opportunity to ask questions.
2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.
3. I give permission for the research interview to be recorded on audio tape.

YES  NO  (please tick as appropriate)

4. I agree to take part in the above study.

\_\_\_\_\_  
Name of Patient                      Signature                      Date

\_\_\_\_\_  
Researcher                              Signature                      Date

1 for patient; 1 for researcher; 1 to be kept with hospital notes

## APPENDIX 5: INTERVIEW TOPIC GUIDE FOR LIPID CLINIC PATIENTS

### Interview topic guide for patients with FH (V3)

1. Introductions and thanks
  - Got PI sheet?
  - Any questions?
  - List of areas, don't have set list of questions, should be quite informal
2. Consent form +**CARBON PAPER**
  - to check everything above board, not pressured into talking with me, understand what participation means
  - Audiotape** OK ?
  - full and accurate record of conversation, my use only, kept confidential and stored securely
3. GIVE COPY OF CONSENT FORM TO PARTICIPANT
4. Start with a little background information
  - Age,
  - what do you do for a living?
  - how would you describe your ethnic background?
  - Who lives in your household?
  - Do you have any children?
5. Can you tell me how you came to be a patient at this clinic?
  - (How did you come to be diagnosed with cholesterol problems)
  - (How did you come to be diagnosed with FH?)
  - When was this?
  - any heart problems?
  
  - family screening, other family members diagnosed, personal history of CHD family history of CHD, raised cholesterol, surprise
6. What were your initial reactions to the diagnosis?
7. Who did you tell about the diagnosis and what did you say?
  - which family members if any and why?
  - Anyone specifically didn't tell?
  - reactions within family, any problems or issues within family
  - other people?
8. What ideas do you have about why you got the heart problem / the cholesterol problem / FH?
  - why then? just come on? Had it for a long time, always? why you?

9. What sort of impact has FH/this condition/the cholesterol problem had on your life?  
Are there any situations where it's caused you difficulty?  
practical, life plans
10. Do you think of it as a serious condition?  
for yourself / for other people  
why / why not
11. In your view, what sort of health problems might people get because of the condition?
12. How do you see your own health in the future?
13. If you had to explain FH/your condition/your cholesterol problem to someone who had never heard of it before, what would you say?
14. What sort of situations do you find yourself talking about FH/your condition/your cholesterol problem and what do you say about it in these situations?  
prospective partner, family, children, work, friends, socialising, other situations
15. Are there any situations where you prefer not to talk about it, or avoid talking about it? (why)  
ever had any bad experiences or strange reactions?
16. What would you say is the worst aspect of having FH your condition/your cholesterol problem?  
Are there any good aspects?
17. Apart from your doctors, are there other places or people you look for information from (or support)?  
sources of information, member of any support group/patients association?
18. It's possible that in the future there might be a specific genetic test for FH/your cholesterol problem. What would you think about that?  
Good? Bad? Any misgivings? Make any difference to you? Your children?