The Potential Value and Regulation of Genetic Tests for Complex Disease Risk Factors

Thesis submitted in accordance with the requirements of the University of London for the degree of Doctor of Philosophy by

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This thesis describes research conducted in the School of Pharmacy, University of London between November 2002 and November 2005 under the supervision of Professor David Taylor and Professor Ian Bates. I certify that the research described is original and that any parts of the work that have been conducted by collaboration are clearly indicated. I also certify that I have written all the text herein and have clearly indicated by suitable citation any part of this dissertation that has already appeared in publications.

__________________________________________
Signature

__________________________________________
Date
This thesis is dedicated to my parents who have supported me in many ways throughout my choices in life.
Abstract

Within twenty years the technology used to identify genetic variations that confer susceptibility to complex diseases is likely to be sufficiently developed so that it can help guide individuals in their efforts to maintain healthy lifestyles. If the knowledge about one’s genetic susceptibility to complex disease risk factors is shown to motivate people to become healthier it is important that this type of information is easily accessible. The current government agenda of promoting individual autonomy and responsibility for health suggests that genetic health risk information should be available in order for the public to use it to inform health-related decisions.

The research presented in this thesis describes the issues discussed during a public consultation exercise on the availability of genetic testing services to the public. It also presents results of an experiment investigating the potential impact of genetic information, and the psychological factors which may influence this impact.

The key findings were as follows:

> Complex disease risk information appears to encourage healthy behaviours, albeit to a greater extent in those already motivated to be healthy, with no apparent negative impact;
> Contrary to some groups’ concerns, stakeholders were not worried that genetic information about complex disease risks would have any negative consequences in terms of employment or insurance discrimination;
> Stakeholders were concerned that the public do not understand well genetic and/or risk information, but it is likely that the public are sufficiently capable of comprehending these concepts;
> Stakeholders showed a ‘cautious shift’ in decision making. This could be related to a reluctance to hand over control of health to the individual or a genuine concern to protect a vulnerable minority of the population.

The thesis discusses the potential impact of regulatory decisions on the principle of increasing individuals’ responsibility for maintaining their health. It argues for a system of access which protects the minority of citizens who are at risk from misinterpreting genetic information and experiencing harm as a consequence, while imposing little or no harm or inconvenience on the majority of citizens.
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How is education supposed to make me feel smarter? Besides, every time I learn something new, it pushes some old stuff out of my brain. Remember when I took that home winemaking course, and I forgot how to drive?

_Homer Simpson_

My greatest thanks are to Professor David Taylor and Professor Ian Bates who not only keep the humour in education but have also been incredibly motivating and supportive. Thanks also to Professor Tony Moffat for his help in the initial stages of the thesis.

I would like to extend my gratitude to all those who gave up their time to be interviewed and to complete questionnaires. Thanks also to all those who assisted in the piloting of the research instruments, and to those at Hewlett Packard, Camden Council, Boots plc and ABPI for distributing questionnaires.

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During the course of the twentieth century the health of the British population improved dramatically. Average life expectancy has increased from around 50 years in 1900 to almost 80 years today. While attempting to cure infections in children and working age adults were among the highest priority tasks of health care professionals at the start of this period, conditions such as heart disease, diabetes, cancers and major mental health problems such as schizophrenia and severe depression have now become the predominant health challenges.

Department of Health publications such as *Choosing health* show that preventing chronic disease occurring or slowing their rate of progression wherever possible is a fundamental objective for the future (Department of Health 2004). Achieving this objective may depend on individuals retaining independence and autonomy and actively choosing better health, rather than adopting a more passive role. Recent initiatives such as the Expert Patient Programme can be taken to reflect a growing acceptance of this amongst policy makers (Department of Health 2001).

Chronic diseases usually have a complex aetiology. They typically result from the influence of many different genetic factors combined with lifestyle factors and environmental influences. Although it may take some years to refine, knowledge about the genetic contribution to health and the development of tests for risk factors related to complex diseases promise to help individuals maintain healthy lifestyles.

However, there are conflicting views about how public access to genetic testing should be regulated and controlled. Some researchers have expressed concerns that uncontrolled exposure to genetic test results could cause needless anxiety amongst some members of the population, bring about fatalism - the perception that a disease is uncontrollable, or false reassurance - the perception that one’s risk is lower if an increased susceptibility-related gene variation is not found (Bates, Templeton, Achter, Harris & Condit 2003; Croyle, Smith, Botkin, Baty & Nash 1997; Lerman, Marshall, Audrain & Gomez-Caminero 1996). For example, an individual told that they have a genetic-based susceptibility to lung cancer may feel that the disease is inevitable and see no benefit in quitting smoking. Or, one told that they do not have a genetic-based susceptibility to lung cancer may feel invulnerable to the disease and therefore see no risk in continuing to smoke.

On the other hand, there is a possibility that a too paternalistic approach to the regulation of genetic testing could prove counterproductive. Excessive degrees of professional control could deny some individuals of convenient access to potentially useful knowledge about their own health. Restricting the access to such knowledge is counter to policy makers’ aims to encourage self care and responsibility for health and disease prevention could be more likely to protect the interest of some health care professionals rather than the public’s health. Should there be concerns about the freedom to access genetic-based health information when there is already freedom to buy tobacco or obtain over-the-counter products whose health effects are sometimes little understood, such as herbal remedies?
This thesis investigates the potential impact of genetic tests for complex disease risk factors, particularly in terms of changing health behaviours. Regulatory issues in this area are explored and the implications of over- or under-regulation of access to genetic information are discussed. It also describes the views and concerns of stakeholders involved in a public consultation about the accessibility of genetic testing to the public. The thesis findings and their implications are discussed and solutions are proposed for the promotion of better public health.

➢ Chapter One presents background information on the impact of gene variations on health and illness, and the wider determinants of public health. It also outlines issues relevant to the regulation of pharmaceutical products and the work of the UK Human Genetics Commission in consulting the public and making recommendations on regulating the availability of genetic tests in the UK.

➢ Chapter Two focuses on the importance of psychological factors on health and illness, and the extent to which they might be effective in changing health-related behaviours. The literature on the interpretation of health risk information is evaluated, including genetic information, and the factors that can influence the understanding of such information. Previous research relating to the impact of genetic health risk information is examined.

➢ Chapter Three sets out the rationale of the thesis and its aims and objectives.

➢ Chapter Four describes qualitative and quantitative research designs and methodologies and how to ensure rigour, credibility and reliability of each.

➢ Chapter Five presents the results of a qualitative analysis of interviews held with stakeholders interested in the regulation of public access to genetic testing and discusses the findings in light of previous research and UK health policy environment. It also presents limitations of the study and suggests some areas for further research.

➢ Chapter Six presents the results of an experimental questionnaire study undertaken with a sample of more than 300 participants. It discusses the results in relation to existing psychological theory and research. Additionally, it states some aspects of the study which could be improved upon and proposes further studies which could rectify these limitations as well as advance the understanding of this area of research.

➢ Chapter Seven highlights key findings from the thesis and draws conclusions based on these. It argues for easily accessible genetic information related to complex diseases and/or risk factors of such conditions. It suggests where such information should be available and proposes a solution to overly cautious regulatory policies.
CHAPTER ONE

GENETICS AND THE PUBLIC'S HEALTH
1.1 Background

This chapter describes the role of genes and gene variations in disease and illness and explains how mutations in DNA occur and how, together with environmental and behavioural factors, they can affect the risk of illnesses such as cancers and cardiovascular disease. Types of genetic tests which are currently in use are described, including those for complex disease risk factors.

The chapter also describes historical changes in public health and follows the recent government agenda of promoting increased individual autonomy over health. It goes on to discuss the potential role of pharmacists in the use of new genetic technology and describes the regulatory issues related to the provision of genetic tests and services to the public. Outlines of the role of public consultation exercises in gathering expert and public opinion and the recent Human Genetics Commission consultation on public access to genetic tests are given.

1.2 The role of genes and gene variants in disease

1.2.1 Gene variations

Genetic mutations, or variations, are changes in a normal sequence of DNA. This can simply be a change in one of the bases or the disappearance or addition of a base. Or, it could be whole segments of DNA or genes which are repeated or have disappeared.

Mutations can either be inherited from parents, or acquired during life. A hereditary mutation is a gene change in the reproductive cells of the parent which is then passed on to the offspring. The mutation is copied every time the cells divide.

Acquired, or somatic, mutations are changes in DNA that arise within individual cells throughout a person’s lifetime. They often occur due to errors in cell division. Some environmental factors can also increase the likelihood of mutation such as radiation (e.g. sunlight) and certain chemicals (e.g. tobacco smoke). DNA repair genes
have the ability to recognise mistakes and correct them before cell division, but these can malfunction or become less efficient with age. Mutations often lead to impairment of the protein production but can occasionally lead to a differently functioning protein, although this usually happens as a result of several mutations. Many mutations have no apparent damaging effect. Some take place in part of the so called ‘junk DNA’ which does not code for proteins. Some mutations are harmful, however, such as those responsible for cancers or cardiovascular disease.

1.2.2 Single nucleotide polymorphisms

Some genetic variations are more common than others. A ‘single nucleotide polymorphism’, or SNP (pronounced ‘snip’), is the most common form of genetic variability in the human genome corresponding to a single nucleotide substitution within a DNA sequence. 99.9% of bases are identical in all human beings, leaving only 0.1% of the genome responsible for all differences between individuals. SNPs account for 90% of this variability and occur approximately every 500-1000 base pairs.

Because we have 23 pairs of chromosomes humans have two copies of each gene. An individual can be either homozygous (having both copies) or heterozygous (only having one copy) for a variation. For example, Figure 1.1 shows a sequence of DNA in three people. The highlighted base in the sequence could be the normal, or ‘wild type’, base (T) or it could be the SNP (G). An individual’s genotype would therefore be one of three alleles: TT (homozygous), TG (heterozygous) or GG (homozygous).

**Figure 1.1: Example of a SNP**

<table>
<thead>
<tr>
<th>Person</th>
<th>DNA Sequence</th>
<th>Genotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>Person 1</td>
<td>C - G - A - T - G - T - C - A</td>
<td>Homozygous (TT wild type)</td>
</tr>
<tr>
<td></td>
<td>C - G - A - T - G - T - C - A</td>
<td></td>
</tr>
<tr>
<td>Person 2</td>
<td>C - G - A - G - G - T - C - A</td>
<td>Homozygous (GG mutation)</td>
</tr>
<tr>
<td></td>
<td>C - G - A - G - G - T - C - A</td>
<td></td>
</tr>
<tr>
<td>Person 3</td>
<td>C - G - A - T - G - T - C - A</td>
<td>Heterozygous (TG mutation)</td>
</tr>
<tr>
<td></td>
<td>C - G - A - G - G - T - C - A</td>
<td></td>
</tr>
</tbody>
</table>
Several SNPs have been associated with increased susceptibility to various diseases, although the evidence is not always conclusive (Ioannidis et al 2001). Table 1.1 shows an indication of the complexity of gene polymorphisms and their association with disease. A gene variant can incur different risks in different sub-populations, such as race or gender. In addition, the same gene variants can be associated with different diseases, such as ApoE4. Similarly, different variants or a combination of variants have been associated with the same disease, such as CYP1A1 and GSTM1.

Table 1.1: Some gene variants found to be associated with disease

<table>
<thead>
<tr>
<th>Gene or gene variant</th>
<th>Approximate prevalence of variant</th>
<th>Disease</th>
<th>Risk status compared with other variations of the gene</th>
</tr>
</thead>
<tbody>
<tr>
<td>APOE e4</td>
<td>Caucasians: 13% Africans: 25% Asians: 10%</td>
<td>Coronary heart disease</td>
<td>Odds ratio of 1.38 (men) and 1.82 (women)(^1)</td>
</tr>
<tr>
<td>APOE e4</td>
<td></td>
<td>Alzheimer's Disease</td>
<td>Odds ratio of 14.9 in Caucasians(^1)</td>
</tr>
<tr>
<td>CYP1A1</td>
<td></td>
<td>Prostate cancer</td>
<td>Odds ratio of 2.38(^2)</td>
</tr>
<tr>
<td>CYP1A1</td>
<td></td>
<td>Breast cancer</td>
<td>Increased risk in female smokers(^3)</td>
</tr>
<tr>
<td>GSTM1</td>
<td>(GSTM1 null) Caucasian: 50% Black: 30%</td>
<td>Bladder cancer</td>
<td>Odds ratio of 1.44 in those with null variant(^4)</td>
</tr>
<tr>
<td>Combination of CYP1A1 and GSTM1</td>
<td>Caucasian: 50%</td>
<td>Lung cancer</td>
<td>Odds ratio of 2.47 in Chinese(^5)</td>
</tr>
<tr>
<td>Combination of CYP1A1 and GSTM1</td>
<td>Black: 30%</td>
<td>Lung cancer</td>
<td>Odds ratio of 4.67 in Caucasian non-smokers(^6)</td>
</tr>
<tr>
<td>Combination of CYP2E1 and GSTM1</td>
<td></td>
<td>Lung cancer</td>
<td>Odds ratio of 3.0 in Japanese(^7)</td>
</tr>
<tr>
<td>MTHFR CT</td>
<td>(homozygous MTHFR CT variation) White: 10% Asian: 4% African American: 1%</td>
<td>Coronary heart disease</td>
<td>Odds ratio of 1.16 in TT genotype(^8)</td>
</tr>
</tbody>
</table>

Table 1.1 is in fact simplified. As indicated above, increased risk of disease not only depends on one gene or one of its alleles but also whether an individual is homozygous or heterozygous for that allele. For example, the ApoE gene has three common alleles: ε2, ε3 and ε4, each producing a different version of apolipoprotein. The various forms of the protein interact differently with lipoprotein receptors, thus altering cholesterol levels. It is the effect on cholesterol levels that link the ApoE gene to coronary heart disease. In simple terms, the ε4 variant raises low density cholesterol levels and the ε2 variant lowers them. Due to having two copies, individuals can have one of six combinations of the gene: ε2/ε2, ε3/ε3, ε4/ε4, ε2/ε3, ε2/ε4 or ε3/ε4. The homozygous polymorphism ε3/ε3 is the most common and ε2/ε2 the least common. Each combination is associated with differing levels of risk for coronary heart disease.

Usually, it takes several variant genes to produce symptoms of a condition. Conditions such as heart disease, cancers, diabetes and mental illnesses such as schizophrenia and depression appear to be caused by complex gene-gene and gene-environment interactions, and are therefore often termed ‘complex diseases’. No one factor alone will cause the disease, although it will affect a person’s likelihood to develop it. A person could carry a predisposition to a disease but may not develop it if environmental triggers are not present. For example, a mutation in the MTHFR gene (for a more detailed explanation of this gene see Section 1.2.4) affects levels of homocysteine, a risk factor for cardiovascular disease. However, the intake of folic acid and other B vitamins affects this relationship. It is therefore much more difficult to predict the occurrence of these types of disease, compared to those caused by single gene mutations.

1.2.3 Genetic influences in cancer

Many polymorphisms of various genes have been associated with increased risk of cancers. To further explain the complex role of gene variants this section describes the GSTM1 gene as an example.

The glutathione S-transferase (GST) enzymes are involved in the Phase II detoxification process. Phase I detoxification involves the ‘activation’ of toxins by
enzymes where molecules of oxygen or nitrogen are attached to the toxin. Phase II
detoxification enzymes then bind the toxin to a chemical called glutathione which
allows the toxin to be dissolved in water, and removed via sweat or urine. The GST
enzymes are important in the metabolism of environmental carcinogens and
chemical toxins and thus the genes encoding them have been linked with cancer
susceptibility.

One polymorphism in the GSTM1 gene ('GSTM1 null') results in it failing to
produce the corresponding enzyme. Around half of Europeans, Asians and Hispanic
people have this polymorphism, along with 29 percent of Africans (Engel et al
2002). GSTM1 has been specifically associated with the detoxification of toxins
found in tobacco smoke. The well-documented risk factor of smoking for bladder
cancer has led to research on the role of the GSTM1 polymorphism in bladder cancer
susceptibility. Engel et al's (2002) meta analysis reported that individuals with the
GSTM1 null polymorphism are at increased risk of bladder cancer, with an odds
ratio of 1.44. In addition, there was a suggestion of an additive interaction with
smoking.

1.2.4 Genetic influences in cardiovascular disease (CVD)

To further explain the role of gene polymorphisms in cardiovascular disease, this
section will use the examples of the ApoE and MTHFR genes.

Apolipoprotein E

The apolipoprotein E gene produces the protein apolipoprotein E (ApoE).
Apolipoproteins combine with free cholesterol to form lipoproteins. ApoE in
particular is critical in the formation of very low density lipoprotein (VLDL) which
deposits cholesterol in the walls of arteries. As briefly described above, the various
forms of the ApoE protein (ε2, ε3 and ε4) interact differently with lipoprotein
receptors, leading to genotypic differences in total and LDL cholesterol levels. In
general, the ε2 allele lowers total cholesterol levels and the ε4 allele raises them. The
ApoE gene contributes more than any other gene to cholesterol variability (Eichner
et al 2002).
High levels of LDL and VLDL cholesterol have been associated with increased risk of CVD. Research has found that e4 carriers have up to 40% increased risk of dying from coronary heart disease compared to e2 or e3 carriers (Eichner et al 2002). Other factors, particularly diet and physical activity, are also known to effect CVD risk, independently and by interacting with genes. ApoE e4 carriers have been shown to respond better to a low fat diet and show greater changes in cholesterol level in response to changes in fat and cholesterol intake (Eichner et al 2002). Other research has shown that the effect of physical activity on LDL cholesterol varies according to ApoE alleles (Taimela et al 1996).

MTHFR
The methylene tetrahydrofolate reductase (MTHFR) enzyme is produced by its respective MTHFR gene. This enzyme is involved in the process of converting homocysteine to methionine. A particular variant of the MTHFR gene, the C677T polymorphism, renders the process less efficient and is correlated with higher homocysteine levels in those who carry it (eg. Andreassi et al 2003). High plasma homocysteine levels have been identified as a risk factor for CVD (Klerk et al 2002).

The MTHFR gene is an excellent example of how genetic polymorphisms can interact with other factors to influence metabolic processes and disease outcomes. Folate is required to convert homocysteine into methionine and high folate intake has been associated with reduced homocysteine levels in the blood. Wald et al (2001) conducted a randomised trial of folic acid supplementation on serum homocysteine levels. They randomised 151 patients with heart disease to one of five folic acid supplement doses: 0.2mg per day, 0.4, 0.6, 0.8, 1.0, or placebo. Serum homocysteine levels decreased with increasing folic acid dosage, although 0.8mg per day achieved similar reductions in homocysteine levels to 1mg/day. The higher the initial homocysteine level, the greater the response to folic acid.

Research has shown that individuals with the homozygous C677T gene variation (TT) are only at higher risk of heart disease if they have low folate status. Similarly, there is no increased homocysteine level, or subsequent increased heart disease risk in TT individuals who also have a high intake of folate (Klerk et al 2002).
The MTHFR example also highlights how environmental factors can counteract the effects of a polymorphism. In one study patients with CVD were given folic acid supplementation (Liu, Chiang & Chen, 2004). After 8 weeks those with the MTHFR TT genotype had decreased their homocysteine levels by 40%, more than any other genotype.

### 1.2.5 Existing genetic technology

There are currently several different types of genetic test available (Table 1.2).

<table>
<thead>
<tr>
<th>Type of Genetic Testing</th>
<th>recessive or dominant gene</th>
<th>Frequency with which disease is developed in individuals who have the gene(s) (&quot;penetrance&quot;)</th>
<th>Single gene or complex gene/environment interaction</th>
<th>Other information</th>
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<tbody>
<tr>
<td><strong>Prenatal diagnostic testing for Down’s Syndrome</strong></td>
<td>Down’s syndrome is caused by having trisomy (3 copies) of chromosome 21.</td>
<td>100%. All babies with trisomy 21 have Down’s Syndrome, but severity of problems can vary.</td>
<td>Caused by duplication of a chromosome rather than a gene mutation.</td>
<td>Down’s can be tested by amniocentesis where a sample of amniotic fluid is taken from pregnant women and subjected to chromosomal analysis.</td>
</tr>
<tr>
<td><strong>Neonatal screening for phenylketonuria (PKU)</strong></td>
<td>PKU is caused by a recessive gene. Having only one copy of the PKU gene variation will result in being a ‘healthy carrier’ of the disease.</td>
<td>Incomplete penetrance. The disease will only present if an individual carries both copies of the gene variation.</td>
<td>Single gene variation.</td>
<td>Neonatal screening for PKU is standard in the UK, although screening actually identifies amino acids in the blood as a result of the gene variation, rather than the gene variation per se.</td>
</tr>
<tr>
<td><strong>Carrier testing for cystic fibrosis (CF)</strong></td>
<td>CF is caused by a recessive gene. Having only one copy of the CF gene variation will result in being a ‘healthy carrier’ of the disease.</td>
<td>Incomplete penetrance. The disease will only present if an individual carries both copies of the gene variation.</td>
<td>Single gene variation.</td>
<td>Currently, the test involves a blood or mouthwash sample. However, neonatal testing for CF will become routine in the UK in the near future in order that babies can have immediate access to screening.</td>
</tr>
<tr>
<td><strong>Predictive tests for Huntington's Disease (HD)</strong></td>
<td><strong>Recessive or dominant gene</strong></td>
<td><strong>Frequency with which disease is developed in individuals who have the gene(s) (&quot;Penetrance&quot;)</strong></td>
<td><strong>Single gene or complex gene/environment interaction</strong></td>
<td><strong>Other information</strong></td>
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<tr>
<td>HD is caused by a repetitive nucleotide sequence on the HD gene. It is inherited in a dominant manner, so that the disease will occur in those who have inherited only one copy of the mutated gene.</td>
<td>Complete penetrance (100%). All individuals with the gene will develop the disease.</td>
<td>Single gene variation.</td>
<td>Predictive testing is available in the UK. Genetic counselling is strongly advised and is intensive. Prenatal testing is less common but is available.</td>
<td></td>
</tr>
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</table>

**Predictive tests for breast/ovarian cancer**

About 5% of breast or ovarian cancer sufferers show mutations in the BRCA1 and BRCA2 genes. The mutations are inherited in a dominant fashion.

Incomplete/variable penetrance. Individuals with the BRCA1/2 mutations have around an 80% chance of developing breast cancer by age 70 (Public Health Genetics Unit, 2005).

Different mutations have been found in different individuals. Breast and ovarian cancers have been associated with the BRCA genes in only 5% of cases (Public Health Genetics Unit, 2005) so 95% of cases are caused by other factors.

Testing for BRCA genes is available on the NHS but is not statutory.

**Susceptibility tests for complex diseases, such as heart disease**

Mutations related to complex disease risk factors can be inherited or can be acquired throughout life.

Incomplete/variable penetrance. Mutations only increase the risk of developing risk factors associated with the disease.

Complex diseases are caused by complex gene-gene and gene-environment interactions.

Involves a cheek swab. Not available on the NHS. Available through some UK and US commercial companies.

**Susceptibility tests for complex diseases**

These tests detect gene variations that increase the risk of developing a complex disease. A swab of cells is commonly taken from the inside of the cheek and submitted for DNA analysis where polymorphisms in one or more genes are identified. A report may be sent to the client or their health professional describing the genetic risk factors.
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the findings and giving environmental and behavioural advice, such as recommended nutrient intake.

Such diseases however, are caused by complex interactions between several genes and also between genes, environment and behaviour. This makes evaluating the risks of disease associated with SNPs difficult and varying between studies as inevitably different population samples are used. Ioannidis et al (2001) conducted a meta-analysis of 370 studies looking at 36 gene-disease associations. They found significant heterogeneity between results of studies looking at the same gene-disease association. The authors suggested that this could have been due to sampling differences. Studies with varying sample sizes or a lack of consideration for different ethnicities, ages, and biological and environmental variability could make studies less reliable and less comparable. Differences in disease expression and disease susceptibility could also explain varying results (Ioannidis et al 2003).

1.3 The changing patterns of public health

This section describes changes in the public health of the UK population and concurrent changes in the predominant health challenges and the nature of health care.

Figure 1.2: Demographic Transition in NW Europe and the US

![Demographic Transition Graph]

- Pre-transitional society, with high fluctuating mortality balanced by high fertility.
- ‘Post industrial’ stage of demographic transition, with aged population and crude death and birth rates low and balanced, starts in the closing decades of the 20th century.
- Start of first phase of transition early industrialisation following agrarian reform in the 1700s.
- Later stage industrialisation begins towards the end of the 19th century.
Figure 1.2 shows how European countries, including the UK, followed by other countries and nations, have moved from a pre-industrialised state characterised by high birth rates and high infant mortality associated with environmental exposure and accidents to one where birth and death rates are low. Between these states, in the UK's Victorian era, birth rates were slowing but not as dramatically as death rates, leading to an increase in population. Now, the UK population would be in a natural decline but for immigration. This demographic transition in the UK was due to economic growth, improved nutrition and hygiene and enhanced education, and only in small part due to medical or pharmaceutical care or availability of medicines or vaccines of any type. Linked to demographic transition is epidemiological transition, the changing patterns of illness and disability over time. As the prevalence of acute or infectious diseases has declined the occurrence and impact of noncommunicable, chronic diseases such as heart disease and stroke has increased.

The term 'transition of care' describes the social changes and new types of health care services which accompany the demographic and epidemiological transitions (Taylor, in press). Following epidemiological transition, Taylor proposes that there is a gradual shift in focus in health care from treatment of illness towards prevention of disease, and the facilitation of individuals with long-term conditions to manage their illness and continue their daily lives as normally as possible. Health professionals' roles now focus more on monitoring chronic conditions, such as diabetes or hypertension, and improving quality of life rather than curing disease. The shift from the paternalistic model of medical care to increased personal responsibility and empowerment also accompanies these changes. Where once the doctor dictated a treatment regimen and the patient was expected to follow it without question, there is now more emphasis on shared decision-making and responsibility for treatment (Taylor, personal communication).

At the same time, as the UK population has become wealthier, better educated, healthier and safer, people are increasingly concerned with risks to themselves and their families' health, illustrated by recent public concerns about mobile phone masts and the MMR vaccination. Additionally, unlike a century ago, many patients are now as well educated as health professionals: this plus modern computer technology
allows patients to access detailed and specialist health information more than ever before, strengthening the involvement of the patient in his or her medical care (Taylor, in press).

Public health has also changed over time. Where once it was concerned with macro level problems such as clean water, it then shifted to micro level problems such as the standard of housing. Recently it has been concerned with individual’s health behaviours and lifestyles, and currently, and in the future, the importance of genetics is emerging.

1.4 Current NHS policies in England

The increasing prevalence of chronic diseases illuminates the significance of health-related behaviours such as smoking, physical activity and diet. Instead of solely relying on health professionals to cure illness, the onus is now on the individual to take some responsibility for their own health. It is apparent that the UK government is in favour of this shift towards individual responsibility. Table 1.3 maps recent government documents which clearly have a common theme of patient or individual autonomy, both in terms of illness prevention and treatment.
Table 1.3: Primary recommendations and conclusions of recent government documents

<table>
<thead>
<tr>
<th>Document</th>
<th>Primary recommendations/conclusions</th>
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<tr>
<td><strong>The NHS Plan</strong>&lt;br&gt;Department of Health&lt;br&gt;July 2000 (a)</td>
<td><em>The NHS Plan</em> is an investment plan for the NHS. Key recommendations include:&lt;br&gt;• Devolvement of power from government to local health services;&lt;br&gt;• More investment in NHS staff and extension of nurse and other staff roles;&lt;br&gt;• Improvement of screening programmes, shorter hospital waiting times, free services;&lt;br&gt;• Patients to have more involvement in the NHS by setting up patient forums, surveys, and advocates.</td>
</tr>
<tr>
<td><strong>Pharmacy in the Future — implementing the NHS Plan.</strong>&lt;br&gt;Department of Health.&lt;br&gt;September 2000 (b)</td>
<td><em>Pharmacy in the future</em> sets out a specific programme of reform as part of the NHS Plan for pharmacists as a key health professional in the NHS.&lt;br&gt;<em>The NHS Plan</em>, published in July 2000 set out to reform the NHS by reducing waiting times, delivering patient-centred quality care, better local services and more emphasis on illness prevention and health maintenance.&lt;br&gt;Priorities centre around pharmacists becoming more involved in the care and treatment of patients by:&lt;br&gt;- Carrying out medication reviews;&lt;br&gt;- Supplementary prescribing (leading to independent prescribing);&lt;br&gt;- Involvement in counselling and support in smoking cessation services.&lt;br&gt;Pharmacists are also key to the government’s investment in the concept of medicines partnerships where patients are empowered to take an active role in their health care.</td>
</tr>
<tr>
<td><strong>The Expert Patient. A new approach to chronic disease management for the 21st century.</strong>&lt;br&gt;Department of Health.&lt;br&gt;September 2001.</td>
<td>Introduces Expert Patient programmes to patients with chronic illness.&lt;br&gt;These self-management programmes teach pain, symptom and stress management, developing coping skills, problem solving, communication with professionals and accessing resources and services.&lt;br&gt;The programmes are not simply about providing education and information to patients but about developing their confidence and motivation to use information, services and their own skills to take control of their lives.&lt;br&gt;They will work in partnership with health professionals to take some responsibility for their treatment and health care.</td>
</tr>
<tr>
<td><strong>Securing our future health: Taking a long-term view.</strong>&lt;br&gt;Wanless&lt;br&gt;April 2002</td>
<td>The Wanless report is an evidence-based assessment of the long-term resource requirements of the NHS. Key recommendations in order to deliver high quality health care in the UK include:&lt;br&gt;• Devoting a significantly larger share of the national income to health care over the next 20 years in addition to using resources more effectively;&lt;br&gt;• More focus and better success in health promotion and disease prevention;&lt;br&gt;• More effective partnership between health professionals and the public by:&lt;br&gt;  - improving health information to help people engage with their care;&lt;br&gt;  - the use of pro-active policies to encourage reductions in health risk factors;&lt;br&gt;  - reinforcing patient involvement in the NHS;&lt;br&gt;  - better understanding of local health services are performing.&lt;br&gt;• Lower resource costs are associated with a more engaged public who live healthier lives and prevent illness</td>
</tr>
<tr>
<td>Document</td>
<td>Primary recommendations/conclusions</td>
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<tr>
<td>The NHS Improvement Plan</td>
<td>The NHS Improvement Plan sets out priorities for the NHS between 2004 and 2008, supporting those stated in The NHS Plan. It summarises the improvements made to the NHS in the preceding years, which include faster and more convenient access to care, better quality care, staff increases and modernisation of facilities. With these improvements in place, the NHS Improvement Plan promises to focus more on: • Better support for people with chronic illnesses by enabling them to manage their own condition in a way that suits them; one of the ways of doing this is to nationally roll out the Expert Patient Programme; • Prioritising disease prevention and health inequalities; • Providing a wider choice of health services to the patient; • Encouraging multidisciplinary teams in primary care.</td>
</tr>
<tr>
<td>Securing good health for the whole population.</td>
<td>• This review focuses on illness prevention and on the cost-effectiveness of interventions to improve the health of the UK population and reduce health inequalities. Key points include: • People need to be supported more actively to make better decisions about their health. Advice should be freely available to assist full engagement of the population. • There is poor evaluation of public health interventions, resulting in a lack of knowledge of what works and for which groups of people. Cost-effectiveness, monitoring and feedback should be an integral part of national programmes. • Objectives should be set for changing the prevalence of important health determinants, particularly those key to reducing inequalities, in order to inform resource planning. • The role of self-care, development of the 'expert patient' and the role of community pharmacists will be developed to expand capacity in managing chronic conditions. • Knowledge of genetics and individual risk factors will have more influence through individualised health promotion and disease prevention.</td>
</tr>
<tr>
<td>Choosing health: Making healthier choices easier.</td>
<td>Vision: • A new approach to public health which “respects the freedom of individual choice in a diverse, open and more questioning society” and “recognises the realities of the impact of the consumer society on those choices”. Promises: • Effective and trustworthy information on health risks via communities, schools and workplaces; • A health information service which will provide easy and confidential access to health information; • The food industry and the media to be involved in providing reliable information; • Develop and improve smoking cessation services; • Tackle obesity; • Engage with employers to improve health in the workplace, including encouraging smoke-free buildings and improving cycle networks.</td>
</tr>
<tr>
<td>Creating a patient-led NHS</td>
<td>• A report on delivering the NHS Improvement Plan. • Key themes are providing a greater choice of services for patients and a change of culture to focus more on health promotion, disease prevention and treating the patient holistically. • In order to do this shared values and codes of conduct, continuing professional learning and clearer leadership will be required.</td>
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<table>
<thead>
<tr>
<th>Document</th>
<th>Primary recommendations/conclusions</th>
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</table>
| **Self-care: A real choice**<br>Department of Health<br>January 2005 (a) | • Provides information to primary care teams, professionals and practitioners about why self-care is important and should be supported.  
• Supporting self-care in individuals leads to improved health and quality of life, increased patient satisfaction and reduced use of health services.  
• Self-care is integral to the NHS key priorities of care of long-term conditions and improved access to, and greater choice of, care.  
• Supporting self-care involves:  
  - accessible advice and information;  
  - self-diagnostic tools;  
  - health education and self-care training;  
  - campaigns on lifestyle issues to change behaviours to promote health and prevent illness. |
| **Choosing health through pharmacy: A programme for pharmaceutical public health 2005-2015.**<br>Department of Health<br>April 2005 (d) | • A pharmacy-specific strategy from the Choosing health White Paper which recognises the importance of pharmacists in improving public health. Areas in which pharmacists can contribute, or can develop, include:  
  - promoting health literacy;  
  - providing smoking cessation, sexual health, immunisation and needle exchange services;  
  - identifying individuals with disease risk factors and offer lifestyle assessments and advice;  
  - contribute to the care of individuals with long-term illnesses by encouraging effective use of medicines, promoting healthy lifestyles, supporting self-care and carrying out medication reviews. |

Genetic testing for complex disease risks will be able to provide individuals with information about the combined effect of their genes and lifestyle on their health. This individually tailored information will allow people to be more informed about the decisions they make regarding their health. The UK government recognised the potential of genetics in health care and published the White Paper *Our inheritance, our future* in June 2003 (Department of Health, 2003b). Its vision was that the NHS would take the lead in maximising the advantages of genetic developments in health care. It acknowledged that over time more would be known about the genetic and environmental contributions to common diseases like heart disease and that testing for predisposition to disease would soon become available. Genetic advances would also allow treatment, lifestyle advice and monitoring to be tailored to an individuals’ genetic profile. However, they also accepted that it would be difficult to predict exactly when these new advances in technology would be exploitable. The importance of public acceptance, confidence and understanding of genetics and related issues was also acknowledged.
Recent evidence suggests that there may be a relatively high public interest in genetic tests for complex diseases. Sanderson, Wardle, Jarvis & Humphries (2004) surveyed almost 2000 UK residents and reported that 69% said that they would definitely or probably take a genetic test for heart disease risk, and 64% said the same for a genetic test for cancer risk. Respondents were asked via a series of questions within a larger omnibus survey; the limited information given to respondents upon which they based their answer is not likely to be reflective of the information they would have available when faced with that situation in real life. For example, many people might look for further details, for example, about costs and medical procedures, on the Internet before making a decision to take the test. In addition, interest expressed in a survey is unlikely to translate into actual uptake of the test in all respondents. Further, 94 percent of the sample was Caucasian, making these results unsuitable for generalisation to other ethnic groups. However, taking these limitations into account it is still possible that a substantial proportion of the UK population may be interested in this information.

1.4.1 A role for pharmacy in genetic testing?
Pharmacogenetics is the use of genetics in the study and clinical use of medicines. SNPs within genes influence factors which affect the absorption and metabolism of some therapeutic agents. This can lead to individual differences in the response to medicines. Some individuals will metabolise drugs slowly, therefore requiring a smaller dose than fast metabolisers. Thus, a standard dose may lead to low or high concentrations in the blood, causing ineffective therapy or severe toxicity, respectively. Examples include 20% of the population who do not respond to beta-blockers and the slow metabolisers who are at risk of toxicity with isoniazid, used in the treatment of tuberculosis (Royal Pharmaceutical Society of Great Britain 2001).

Currently, genetic variations related to the processing of medicines are not considered when prescribing treatments. However, the genetics White Paper recognised the potential that pharmacogenetics can have on targeting medicines more effectively, improving patient outcomes, reducing adverse side effects and producing significant savings for the NHS, although in reality it will not enter mainstream healthcare for 15 to 20 years (Department of Health 2003b; Royal Pharmaceutical Society of Great Britain 2001; The Royal Society 2005). It is
envisaged that pharmacists will take on a greater role in prescribing, where, with the aid of genetic testing, they can select the most favourable treatment option for the patient.

Pharmacists’ roles in the care and treatment of chronic diseases and involvement in illness risk-reducing services such as smoking cessation and cholesterol testing (as proposed by the *Choosing health through pharmacy* strategy) also allows them to have a great influence on public health. The emergence of new policies such as these for enhanced pharmaceutical services together with pharmacists increasing knowledge of genetics supplementing their education and training background, the improved technology in community pharmacies and their accessibility to the public makes them well placed to deliver genetic testing services and give related advice. However, there has been considerable debate over where genetic testing services should be accessed, as discussed in Section 1.5.3.

### 1.5 Regulating for public protection and health improvement

The aim of medicines-related regulation has changed over time, and can be linked with epidemiological transition discussed earlier in this chapter. Initially it was introduced to control the sale of poisons to the public. In 1956 the drug thalidomide went on sale in Europe. It was prescribed as a treatment for vomiting in early pregnancy and proved popular due to its efficacy. However, a dramatic increase in the number of malformed babies was seen in the early 1960s and thalidomide was found to be the cause. Prior to this medicines regulation was relatively reactive – it was the industry’s decision to put drugs on the market and the state intervened only when adverse events were apparent. For example, the quality of vaccines was poorly guaranteed and a lack of standardisation involved no accurate measure of medicine purity or effectiveness. The UK reacted to the thalidomide incident by immediately establishing a Committee on the Safety of Drugs which came into operation in 1963 (Tucker & Taylor 2001).

When the Labour government came into power in 1964 they sought to expand high technology industries like pharmaceuticals whilst recognising the need to regulate in
order to regain public trust in the wake of the thalidomide tragedy. The Committee on the Safety of Drugs was seen as ineffectual as it relied on the voluntary participation of the industry. The 1968 Medicines Act instigated the formation of advisory bodies including the Medicines Commission, advising Ministers on broad aspects of medicines policy, and the Committee on the Safety of Medicines. The Act also introduced the medicine licensing system, which still exists today, based on three categories of prescription only medicines (POM), pharmacy medicines (P) and medicines on a general sales list (GSM). All medicines were now to satisfy the licensing authority of their quality, efficacy and safety.

The pharmaceutical industry has grown in the last 30 years to be one of the most profitable industry in Britain. There have recently been criticisms that the power of this industry has developed at the expense of public interest (House of Commons Health Committee 2005). Recent controversies over the side effects of drugs such as Vioxx (an arthritis drug withdrawn from the market due to links with heart attacks and stroke: Singh 2004) and some selective serotonin reuptake inhibitors (a type of antidepressant drug which has been linked with suicide attempts: Fergusson et al 2005) due to inadequate trials and post-marketing surveillance procedures echo that of thalidomide and have highlighted the increased complexity of drug technology and the failures of regulatory agencies.

In conjunction with the new genetic technology a regulatory framework is required to ensure that it is used to its best advantage. There are many areas of genetic testing which require regulation, including manufacturing standards, laboratory quality and control, advertising standards, counselling and the use of genetic information for insurance and employment discrimination. This thesis concentrates on the regulation of public access to genetic testing.

1.5.1 To protect or to expose?

Historically, regulation has been used to control monopolies and promote fair market competition. During the 1960s and 1970s there was a growth in ‘social regulation’ - using regulation to protect consumers from risks. Debate has ensued on whether the state should protect its citizens by setting out conditions which determine how a
product may be used or by providing them with information about risks in order to evaluate a product or activity for themselves.

Risks can be limited by specifying the standards and quality of products. Avoiding deaths and disabilities caused by drugs or medical devices which have been inadequately trialled and monitored is an obvious reason for restrictive regulation. Some believe that genetic tests can be harmful and should therefore be rigorously regulated (eg. Consumers Association 2003). For example, research has indicated that some patients who have undergone genetic tests for cancer show increased depressive symptoms (Lerman et al 1998). There is also concern that knowing one does not have genetic variations associated with cancer can cause some people to feel invulnerable (eg Lerman et al 1996). However, findings on the emotional and cognitive effects of genetic test results are not conclusive (see Section 2.1.5).

Despite the obvious advantages of protective regulation there are several arguments against it. A framework which restricts the availability of products established at one point in time may quickly become out of date. This may be particularly true in the context of genetic testing. This is an area that, at present, is relatively new. Regulations developed now are likely to be unsuitable in twenty years time when both the technology and the concept are better established, understood and accepted.

It could also restrict the freedom to choose products and the range of choices available. Others argue that regulation can restrict economic innovation and stunt entrepreneurial growth which may force industries to move to unregulated markets. This may be relevant in the area of genetic testing where strict regulation may inhibit the success of new companies and drive them to do business in countries such as the US where health care is much less regulated.

Imposing regulation may exaggerate the severity or likelihood of the risk being regulated. Focussing on risks also detracts from the benefits, and strict regulation may remove products with benefits from the market. In this context, regulation may encourage the notion of ‘genetic exceptionalism’, where genetic tests, information or conditions are treated differently, usually more negatively, than other medical information. By focussing on the negative risks of genetic testing such as
psychological harm, the potential for the genetic information to encourage healthier behaviour is disregarded. The balance of risks and benefits of the product must be considered, both of which can be highly subjective, making this type of regulation controversial.

Failure to understand the limits of a protective approach to regulating genetic testing could lead to a paternalistic approach, which may be counterproductive if it prevents individuals from accessing potentially useful knowledge about their health. Camerer et al (2003) have suggested that a legitimate solution called ‘asymmetric paternalism’ which neither benefits nor harms the majority of ‘rational citizens’ yet protects the minority likely to be at risk.

1.5.2 The role of consultation exercises in gathering opinion

Public consultation exercises are designed to gather expert and public opinion on a specified topic in order to make recommendations for policy and regulation. Historically, policies were based on government and expert opinion. However, in 2000, the House of Lords Select Committee on Science and Technology published a report which argued that public involvement in science-based policy making should be mandatory rather than a bonus (House of Lords Select Committee on Science and Technology 2000). As the BSE crisis and concerns over GM foods had reduced public confidence in scientists, the government and regulatory mechanisms, the Science and Society report emphasised increased transparency regarding scientific advice, the recognition of scientific uncertainty and the legitimacy of public values and concerns (Irwin 2001).

Public consultations are perceived as a fair way of deciding on regulations although interesting issues have been raised (Irwin 2001). For example, are public opinions given the same importance as scientific opinions? What happens when public opinion is in opposition to government policy?

Previous research has shown that despite good intentions, the design of public consultations can be restrictive and may prevent discussion of the topic within the context of everyday life (Irwin 2001). Structured formats of focus groups or surveys
can initiate reactive discussion but may preclude more active debate. Additionally, the format of the consultation specifically described in Irwin’s (2001) article on developments in the biosciences was said to be “very much shaped by the governmental sponsors, the advisory group and the researchers” (p.13).

Another potential issue in consultation exercises is how and whether stakeholders such as patient groups or professional bodies give a view representative of their organisation. Does each organisation conduct a mini-consultation of its members before providing an official response?

In addition, when responding to a consultation on a scientific issue, are stakeholders more cautious in their recommendations? Social psychology research has shown that group decisions about risk taking can be more cautious than decisions made individually (Stoner 1968), possibly due to perceptions of the widely held opinion (Eliaz, Ray & Razin 2004). Recent controversies around GM foods or the possible link between the MMR vaccination and autism may affect judgement of policy makers. Moreover, genetic testing is under the biotechnology umbrella. Both policy makers and the public may associate features or fears of other types of biotechnology, such as eugenics or cloning, with genetic testing. This may affect decision making. Additionally, the ‘deficit’ model of public understanding of science is that the general public is scientifically illiterate and that this ignorance contributes to their resistance to new technologies (Sturgis, Cooper & Fife-Schaw 2005). Do stakeholders make recommendations for over-regulation because they believe the public do not understand scientific developments?

1.5.3 Human Genetics Commission consultation

In 1999 the Human Genetics Commission (HGC) was established to take on the responsibilities of three previously established government advisory committees, which included the role of providing advice and guidance to ministers on the regulatory framework controlling public access to genetic tests. In February 2002 the HGC was charged with reviewing the supply of genetic testing services direct to the public and making recommendations on how this area should be regulated. A consultation document was published in July 2002. It invited opinions from a broad range of interested parties, from consumerists to patient organisations to industrial
bodies. The HGC held focus groups and a series of public meetings, and conducted an internet-based opinion survey. In order to make recommendations on where genetic tests should be available, the HGC considered several issues related to the public access of genetic testing services (Box 1).
Box 1: Some of the issues discussed during the Human Genetics Commission consultation on genetic tests supplied direct to the public.

**Uniqueness of genetic tests**
What distinctions are there between genetic tests and other health-related tests? Should genetic tests have restricted availability if there are already non-genetic tests available to the public?

**Right to access information**
Should individuals be entitled to obtain genetic information in whatever way they wish?

**Analytical validity, clinical validity and clinical utility of genetic tests**
'Analytical validity' refers to how consistently the test measures what it purports to measure, 'clinical validity' is the accuracy with which the test predicts the presence of absence of a condition or predisposition, and 'clinical utility' relates to the value of the test results for the individual - how useful and informative the test is. Should there be regulation or should market forces determine these standards?

**Security, storage and confidentiality of samples and data**
For how long are DNA samples and data kept at a laboratory? How traceable to the individual is that information? Should laboratories keep data for research purposes? Should family members be informed of genetic test results? Should genetic information be stored on GP records? Genetic test results may have important implications for the future healthcare of a patient. However, this information would then be available to insurance companies or employers.

**Therapeutic gap**
Should genetic tests be available when there is currently no treatment for the illness or disease they test for?

**Counselling**
Should purchasers of genetic tests for complex disease risks be required to undergo genetic counselling? If so, by whom and how extensive should that counselling be?

**Insurance and employment discrimination**
Should genetic information be available to insurance companies to calculate life insurance policies? Should employers be allowed to access genetic test results, or insist on potential employees taking genetic tests in order to make employment or promotion decisions?

**Education of health professionals**
Are health professionals in the UK sufficiently informed and educated about genetic tests and their implications for health?

**Advertising**
How should adverts for genetic testing services be controlled, if at all?
Another factor the HGC had to consider when discussing regulation of access was the type of genetic test. Different tests provide information which has different levels of impact on the individual. At the time of the consultation there was relatively little evidence on the impact of genetic tests for complex disease risk factors.

The consultation document asked responders to bear these issues in mind when considering the regulation of genetic testing services direct to the public. Four options of regulation were suggested by the HGC. The first was no specific regulation. In this option, market forces would essentially determine the quality and reliability of tests. Professional and business codes of ethics and broader frameworks of commercial and consumer legislation would still stand, as would current controls on advertising. Existing products with this level of regulation include home pregnancy testing kits.

The second option was voluntary regulation. Providers of genetic tests would have to adhere to a voluntary code of practice in addition to existing general frameworks. Codes of practice could be introduced by the government, an advisory body or the industry. It would not be illegal to fail to abide by the code but it is likely that there would be commercial consequences.

The third option was voluntary regulation with restrictions on the type of tests that can be offered, which is essentially the current position in the UK. So, tests that may have a high impact on an individual (a test for Huntington’s disease for instance) or which would require advice from genetic counsellors or medical practitioners would not be allowed to be sold directly to the public. Tests would have to be classified into those which could and could not be sold directly.

Strict regulation was the final option. This was similar to the third option except that it might be an offence for a person other than a registered medical practitioner, or other authorised person, to offer a genetic test.

In essence, this chapter highlights the increasing significance of genetics in public health in the twenty first century. It also shows the value and hazards of under- and over-regulation of access to genetic information. The following chapter explains the
role of behaviour in health and illness, and the importance of psychology in changing behaviours to improve health and prevent disease. In addition, it discusses and evaluates evidence relating to the interpretation of health risk messages and the subsequent impact of such information.
CHAPTER TWO

PSYCHOLOGY AND CHANGING BEHAVIOUR
2.1 Psychology and changing behaviour

This chapter focuses on the importance of behavioural factors in health and illness, and the importance of psychology in changing these behaviours. The literature on the interpretation of health risk information is evaluated, including genetic test information, and the factors that can influence the understanding of such information. The chapter also discusses previous research relating to the potential impact of genetic health risk information.

2.1.1 The Importance of behaviour in health and illness

At the end of the nineteenth century the medical model and the 'germ theory' of disease (the theory that disease is caused by pathogens) prompted research into the discovery of pathogens and respective treatments. Such research subsequently led to the development of new medicines, vaccines, technology and surgical procedures which in turn brought about great advances in health such as the prevention and treatment of malaria, tuberculosis, measles, influenza and smallpox.

The elimination of infectious disease allowed better health and longer lifespans. However, this meant that noncommunicable diseases such as cancer and heart disease had time to develop and were exposed as the new biggest killers. Cardiovascular disease, diabetes, cancer and obesity account for nearly 60% of deaths worldwide. The physiological aspects of most diseases are now well known. However, research has also shown that the diseases of today demand an understanding of psychological, social, cultural and behavioural factors. Eighty percent of cases of coronary heart disease, ninety percent of Type 2 diabetes and one third of cancers can be avoided by a healthier diet, increased physical activity and stopping smoking (World Health Organization 2003). Not only are behaviours implicated in the majority of morbidity and mortality in the world, but in turn social, emotional, cognitive and psychological factors are strongly linked to health behaviours (Figure 2.1). Instead of the traditional medical model of illness a fully integrated understanding of disease is now required which involves many disciplines.
2.1.1.1 Health behaviours

Smoking

Smoking is the biggest single cause of preventable illness and premature death. Half of all long term smokers will die of a smoking-related illness. Around 120,000 people in the UK are killed every year due to smoking. It causes thirty percent of all cancer deaths and one out of seven deaths from heart disease. In addition to these
health costs, treating smoking-related illness and disease costs the NHS £1.7 billion per year (Department of Health 1998).

**Diet**

Diet is thought to contribute to one third of all cancers (Department of Health 2000c) and has also been linked to coronary heart disease, stroke, asthma and diabetes. Obesity has been linked with cancers of the oesophagus, colorectum, breast and kidney and the risk of such cancers may be lowered with increased intake of fruit and vegetables (Key, Allen, Spencer & Travis 2002). The UK government recently launched a programme aimed at raising awareness of the benefits of fruit and vegetables in the diet, and improving access to such foods. The ‘5 A Day’ initiative was launched in 2003 and recommends that everyone should eat at least five portions of fruit and vegetables per day.

**Physical activity**

Only 37% of men and 24% of women currently meet the minimum recommendation for physical activity and are sufficiently active to benefit health (Department of Health 2005b). Physical activity is protective against diseases such as coronary heart disease, osteoporosis, depression and anxiety. Sedentary and unfit people have almost double the risk of dying from coronary heart disease and inactivity contributes to the 22% of men and 23% of women who are obese. As part of the Choosing Health White Paper the Department of Health recommends that adults should achieve a total of thirty minutes of at least moderate intensity exercise on at least five days per week (Department of Health 2004b). To prevent obesity, it is recommended that 45 to 60 minutes per day is required.

Quitting smoking, changing eating habits and increasing physical activity is not straightforward however. As mentioned above, psychological research has shown that social, emotional and psychological factors all interact to influence health behaviours.
2.1.2 The role of psychology in understanding health behaviour

The discipline of health psychology seeks to understand the psychological influences on health and illness. Specific areas of research include understanding the factors which influence well-being, developing techniques for prevention and treatment of illness and maintenance of health, and seeking to understand health- or illness-related behaviours. Many theories and concepts have been suggested and evaluated in relation to health behaviours, some of which are described below.

Health behaviour theories

Health behaviour theories, models and concepts have been developed over the years in attempts to explain and predict health behaviours. Examples of extensively researched behaviour models include The Health Belief Model (HBM: Rosenstock 1966) and The Theory of Planned Behaviour (TPB: Ajzen 1985).

The Health Belief Model is a health specific social cognition model whose key components consist of: perceived susceptibility to disease or illness; perceived severity of the disease or illness; perceived benefits of a particular behaviour which offsets the risk of disease or illness; perceived barriers to performing that behaviour or action; cues (reminders or prompts) to take action to reduce illness risks; and, demographic and socio-economic variables. Since the late 1970s Bandura’s concept of self-efficacy (see Bandura 1986) has on occasions been added to the HBM. Self-efficacy (also see below) is the belief an individual holds in relation to his or her ability to undertake the behaviour required.

Although it has been, and is still being, used in many areas of health and illness the HBM has a weak ability to predict health-related behaviours. A meta-analysis of studies using the HBM in adult populations found weak effect sizes of the HBM components in relation to health behaviour, accounting for up to only 9 percent of variance (Harrison et al 1992). It can also be accused of assuming that individuals are rational and logical in their behavioural decision making, and thus health behaviours are under volitional control. No account is taken of unconscious decisions, habits, or emotional or social influences. In addition, the HBM is rarely
used proactively to design behaviour change interventions, probably due to the lack of combinatorial rules regarding its constructs.

The Theory of Planned Behaviour describes 'social cognitions' – an individual's representation of their social environment. These are represented in the model by attitudes and subjective norms. General positive or negative attitudes towards the behaviour are determined by the antecedent beliefs about the behaviour, linking it to certain outcomes. Subjective norms are the perceptions of general social pressures to perform behaviour. This is determined by the antecedent beliefs that important individuals or groups approve or disapprove of the behaviour. The concept of Perceived Behavioural Control (PBC) in the model reflects the individual's perceptions of external and internal factors which may help or hinder the behaviour, and the confidence with which an individual believes he or she can perform the behaviour taking into account such factors. The TPB holds that the immediate precursor to behaviour is behavioural intention, and that beliefs, attitudes and subjective norms predict this rather than behaviour directly (with the exception of PBC which is thought to directly influence behaviour as well as behavioural intention).

The TPB has been successful in predicting a variety of health behaviours, in addition to other behaviours such as public transport use and recycling (Anable 2005; Davis, Phillips, Read & Iida 2006). For example, Hausenblas, Carron & Mack (1997) conducted a meta-analysis on studies using the TPB with exercise behaviour which revealed large effect sizes between attitudes, intention, PBC and exercise behaviour. However, they found low or no relationship between subjective norms, intention and behaviour. Similar results were reported by Armitage & Connor (2001) who conducted a large meta-analysis of TPB studies on several behaviours. Although the TPB accounted for 27 per cent of variance in behaviour (reduced to 21 per cent if observed behaviour rather than self-reported behaviour was used as an outcome) the subjective norm construct was found to be a relatively weak predictor.

Some researchers have suggested ways to increase the predictive power of the TPB. Authors of a recent study investigating intention to increase physical activity suggested that affective determinants, such as likes and dislikes of a particular
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behaviour, should also be considered in addition to instrumental determinants such as advantages and disadvantages (French, Sutton, Hennings et al. 2005). In addition, Abraham et al (1998) suggested individuals take part in activities as a way of preparing themselves to implement the desired behaviour.

The TPB attracts the same criticism as the HBM that it presupposes rationality in the individual. Indeed, the assumption that behaviours are intended is reflected in its name. The inclusion of PBC into the TPB could be said to be a recognition that its predecessor, the Theory of Reasoned Action (Fishbein 1967) was inadequate in predicting behaviours which had strong external influences.

Both models, and indeed other (social) cognition models such as the Protection Motivation Theory (Rogers 1975) and the Health Action Process Approach (Schwarzer 1992), can also be criticised for failing to adequately incorporate social, economic and environmental influences of behaviour. Yarborough & Braden (2001) found that observed variance in breast cancer screening behaviour was higher when socio-economic status was included with HBM components. One could argue that PBC takes into account known influences but this is not specified. The addition of social, economic and environmental predictors of behaviour to such models is likely to strengthen their predictive power.

Self-efficacy

The concept of self efficacy, mentioned briefly above, evolved from Rotter’s social learning theory in the 1950s which held that in order to carry out a behaviour an individual must value the outcome of the behaviour in addition to having an expectancy that the behaviour will produce that outcome (Rotter 1954). So, for example, for an individual to quit smoking he or she must value their health in addition to expecting that quitting smoking will make them healthier.

Bandura developed a separate social learning theory, naming it ‘social cognitive theory’. He explicitly distinguished the concept of outcome expectancy from the concept of self efficacy - individuals’ “judgements of their capabilities to organise and execute courses of action required to attain designated types of performances” (Bandura 1986, p.391). One can have high outcome expectancy (e.g. that quitting
smoking will improve one’s health) yet have low self efficacy (e.g. I do not feel that I am capable of quitting smoking).

Self efficacy is understood to encompass the execution of behaviours and the perceived strength of the ability to regulate motivation, thought processes, affective states and the social and physical environment. Self efficacy can be influenced by past experience, observational or vicarious experience, verbal feedback, physiological states, affective states and the environment. Its important contribution to health behaviour is reflected in its inclusion in the majority of cognitive and social-cognitive models of health behaviour: the Health Belief Model, Protection Motivation Theory, Health Action Process Approach and the Theory of Planned Behaviour. Ajzen (1991) argued that the Perceived Behavioural Control component of the TPB is identical to the concept of self efficacy. However, Bandura (1992) maintains that the two concepts are different. He holds that self efficacy relates to internal (cognitive) control factors whereas perceived behavioural control reflects more general, external factors. To date there is equivocal evidence for the distinction (Armitage & Connor 2001).

As well as influencing the impact of therapeutic interventions, responses to health communications, adherence to health behaviours, relapse to addictive behaviours, recovery after illness and the level of functioning in patients with chronic illness, self-efficacy has been shown to influence the likelihood of initiating health-related behaviours (DeVellis & DeVellis 2001).

Locus of control

The theory of locus of control is rooted in attribution theory which holds that individuals like to attribute causes to events in the world in order to be better able to predict and control it (Kelley 1967). Causal attributions about behaviour have four dimensions: they can be internal or external to the individual, stable or unstable over time, global or specific to situations, and controllable or uncontrollable by the individual.

The internal-external dimension of attribution theory has been specifically applied to health and a measure of health locus of control has been developed (Wallston,
Wallston & DeVellis 1978). This evaluates which individuals are proposed to attribute personal responsibility (internal) for their health-related outcomes or behaviour and which attribute responsibility externally to chance or powerful others (external), such as doctors or family members.

Many studies have investigated health locus of control with the assumption that individuals with a high internal locus of control are more likely to engage in health promoting or risk reducing behaviours. It has been used in studies investigating behaviours such as changing diet, physical activity and adherence to medication, as well as physical and psychosocial functioning (O’Hea et al 2005; Schroder & Schwarzer 2005; Triemstra, Van der Ploeg, Smit, Briet, Ader & Rosendaal 1998). In the field of genetics it has been recently used to investigate the causal attributions of bipolar disorder and interest in genetic testing for the illness (Meiser, Mitchell, McGirr, Van Herten & Schofield 2005).

However, this association between internal locus of control and performance of healthier behaviour has only been partially supported. Steptoe & Wardle (2001) examined locus of control beliefs and ten different health behaviours in over 7000 young adults from 21 European countries. Yet while the associations between internal locus of control and dental health behaviour were consistent, those between locus of control and smoking, physical activity, alcohol consumption and healthy food choices tended to be inconsistent.

They also found, like previous studies, that the correlations between locus of control and behaviours were small and accounted for little variance. However, when applying relative risk analyses they found more promising results. The odds of carrying out healthy behaviours were 40-70% greater in those in the highest quartiles of internal locus of control compared with the lowest quartile. Moreover, individuals scoring high on chance locus of control were 20-35% less likely to carry out healthy behaviours. They conclude that health locus of control is a generalised construct relevant to health and as such may be less precise in predicting health behaviours than more specific variables such as behavioural intention. However, it can denote individuals’ dispositions towards healthy lifestyles.
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Health value

Health value was a concept integral to Rotter's (1954) social learning theory described above. Later, Wallston (1992) argued that internal locus of control will have a stronger influence on health behaviour among individuals who value their health highly compared with those who value it below other priorities. It is predicted that individuals with high health value are more likely to perform healthy behaviours.

However, this relationship is not necessarily straightforward. Researchers have shown that health values directly predict adolescents' intention to use condoms with casual partners, even when controlling for demographic variables, social desirability, history of sexually transmitted infections, previous condom behaviour and other predictors from the Theory of Planned Behaviour (Rosengard et al 2001). Yet health value did not predict condom use with main partners. This may be because the health threat from longer-term partners is perceived to be less and the maintenance of the relationship is more important than health. The study also reported that health value moderated the relationship between attitudes towards condoms and intentions to use them. Adolescents who placed a high value on health have a high intention to use condoms regardless of their attitudes. Conversely, attitudes play a greater role in determining condom intentions when health value is low.

Self-rated health

Individuals' perception of their own health status has been shown to be a powerful predictor of actual health, despite the variation in the cognitive process individuals may go through to reach a conclusion to the question. Idler & Benyamini (1997) reviewed 27 studies of associations between self-ratings of health and mortality. They found that all but four studies showed an independent effect when all other variables such as medication use, health care utilisation, symptoms, physiological measures and smoking practices were considered. Results almost always showed a dose-response effect, such that the highest probability of death is for the category of poorest health.

The authors suggest several reasons for this finding: that self-rated health is more inclusive and accurate than other variables studied, capturing past illnesses,
undiagnosed symptoms and family history; or, that self-rated health may reflect personal resources such as coping strategies or emotional state, and external resources, such as social support or income which can affect mortality. They also suggest that self-rated health may influence health-related behaviours which subsequently affect health status. Indeed, smoking and physical activity have been found to be significantly associated with self-rated health in a Danish study (Moller, Kristensen & Hollnagel 1996).

Self-efficacy, health locus of control, health value and self-rated health have all been shown to be related to the performance of health behaviours, albeit not conclusively or clearly. To date there has been no research on the influence or effect of these concepts on the behavioural impact of genetic information relating to the relationship between genetic variations, folic acid, homocysteine and heart disease risk. It is for exploratory reasons that these concepts are included in the research described in this thesis. One would hypothesise that increased intention to change behaviour would be related to higher self-efficacy, an internal health locus of control and a high health value.

The following two sections discuss how the interpretation of health information can vary according to individual differences and how details within the message can affect its impact.

2.2 Influences on the interpretation of health information

2.2.1 Individual differences

Health (risk) information can also be interpreted differently by individuals which can then lead to differences in behaviour. Recently, a cognitive-social health information processing (C-SHIP) model has been developed which highlights areas in which individuals differ in the way they cope with health and disease threats (Miller, Shoda & Hurley 1996). The model proposes that there are several psychological variables (such as beliefs, emotions and values) involved in the processing of and responding to health information. These factors interact with each other to determine decision
making, cognitions and behaviour related to the illness threat (Miller, Fang, Diefenbach & Bales 2001).

Individuals differ in the activation and processing of these variables but there is evidence to suggest that there are two primary attentional styles which can be identified: ‘monitor’ and ‘blunter’ (Miller 1987, 1995). Broadly, ‘monitoring’ involves actively seeking out and amplifying threatening information whereas ‘blunting’ involves distraction from, and minimising, threat information. A recent review by Miller et al (2001) describes how monitors and blunters differ in the constructs of the C-SHIP framework:

1) Appraisal of susceptibility to illness and disease
Monitors tend to seek out, focus on and amplify threatening information. As a consequence, they tend to overestimate their perceived risk of developing a disease which may adversely affect the uptake of preventative behaviours (Lerman et al 1993; Schwartz et al 1995).

2) Beliefs and expectations about illness prevention/ treatment and one’s ability to perform behaviours to prevent/treat the illness.
Monitors are more likely to have negative expectations about the seriousness and consequences of a health threat and are less likely to believe that there will be an effective treatment or cure for the illness than blunters (Miller et al 2001). They are also more likely to blame themselves for their illness and tend to have lower self-efficacy. This profile disposes monitors to hold pessimistic opinions about their illness and treatment which may increase the stressfulness of the experience and reinforce negative perceptions.

3) Affective and emotional states triggered by a health threat such as anxiety, fear, anger and avoidant thinking.
Research has shown monitors to display higher levels of anxiety, distress and concern than blunters when faced with a health threat, which can persist over time (Miller et al 2001). These affective reactions may lead to a decrease in effective coping and risk-reducing behaviour.

4) Valued and desired health outcomes and their subjective importance and intention to achieve health-related goals.
Monitors tend to desire and seek out information, and more detailed information, about health and illness compared to blunters. Blunters on the other hand are more satisfied with minimal information about their illness, its aetiology, medication side effects and preventative measures. Monitors may therefore be dissatisfied with information given to them by health professionals and this may increase levels of anxiety.

5) Knowledge and strategies for maintaining critical health behaviours or dealing with barriers to health behaviours.

The C-SHIP framework predicts that two skills are important to successful coping with a health threat: a) the ability to plan effectively, such as attending cancer screenings, and b) the ability to manage distress and anxiety related to the threat. Monitors have been found to be more likely to adhere to recommended risk-reducing behaviours due to their higher attention to threats. Conversely, because blunters tend to minimise threat information, they are less likely to perform such behaviours. However, the situation may change when faced with a high level of threat. It is thought that due to the tendency for monitors to become anxious, they may become overly distressed and as a consequence, actively avoid reminders of the threat, such as risk-reducing behaviours, which can lead to denial and disengagement.

In relation to genetic testing, one study found that monitors reported more anxiety before and after counselling for hereditary cancer, than non-monitors (Nordin, Liden, Hanssen et al 2002). However, this result differed according to the instrument measuring monitoring and blunting. Higher monitoring has also been associated with greater psychological distress while anticipating genetic test results for the BRCA1 and BRCA 2 mutations (Tercyak, Lerman, Peshkin et al 2001). High perceived risk among women at risk for ovarian cancer who scored highly on monitoring was also found by Schwartz, Lerman, Miller et al (1995).

To date, Miller's theory of differing attentional coping styles have not been examined in relation to the interpretation of information relating to the relationship between genetic variations, folic acid, homocysteine and heart disease risk.
2.2.2 Message content, framing and delivery

In addition to individual differences subtle details within the message can affect the way the information is received and the message understood.

Type of information

Research has shown that other ways of presentation can have differing effects. Risks presented as relative (e.g. a higher/lower chance than the average person), rather than absolute (e.g. 40% chance) can be more salient (Edwards et al 2001). Risks shown as frequencies (e.g. 40,000 people per year get this disease) rather than probabilities (e.g. 1 in six people get this disease) can be better understood, although this does not necessarily translate into meaningful behaviour (Wilkniss 2000). In addition, presenting a higher number of adverse outcomes (e.g. 300 out of 10,000 compared to 3 out of 100) can have a greater effect (Klein 1997; Yamagishi 1997).

It is likely that other factors such as individual experience and perceptions, and frequency of the target behaviour (a one-off vaccine versus monthly breast self-examination, for example) will also affect the efficacy of a message and therefore the performance of a behaviour.

Some research has found that individually tailored risk and behaviour change information, as opposed to general risk information, prompted more behaviour change (Kreuter & Strecher 1996). However, this applied to cholesterol screening, dietary fat intake and physical activity but not smoking, mammography or Pap smears. Other research has found that genetic information is no more likely to promote behaviour change than any other information (Lerman et al 2000).

Amount of information

The provision of differing amounts and types of information can impact on an individual on many levels: emotional, cognitive and behavioural. The provision of more risk information has been associated with improved patient knowledge and satisfaction but without an increase in anxiety (Garrud, Wood & Stainsby 2001). Some patients want information on other patients who have coped better with the disease (Bennenbroek, Buunk, van der Zee & Grol 2002), and others rate
information on risk factors as more important than information about anatomy, medication or psychological factors (Scott & Thompson 2003).

There is conflicting evidence on the relationship between information needs and emotional state. Kaplowitz, Campo & Chui (2002) found that cancer patients who were more anxious requested less information whereas results from a different study showed that cancer patients' information needs were related to higher levels of anxiety (Mesters, van den Borne, De Boer & Pruyn 2001).

In terms of behavioural impact, one review found that manipulations in amount of information, graphical illustrations of risk and information based on real patient cases (rather than on a population) were not successful in improving desired behavioural outcomes although some manipulations appeared to increase knowledge and affect treatment choices (Edwards et al 2001).

However, needs are likely to vary between individuals. Duggan et al (2002) found that some individuals did not want extensive information about their prescribed drugs and that detailed information would worry or frighten them. Some patients however, wanted more information and believed that specific details about their medication would help them in their self care. This could be related to the monitor/blunter coping styles but this was not investigated. Other research has found that desire for information is related to socio-economic variables (Borja Lopetegi et al 2002). Individual health beliefs and perceptions may also influence the amount of information desired. One study showed that patients who perceived prescription drugs as beneficial were more likely to desire more information about their medicines (Laaksonen, Duggan & Bates 2002).

It has also been suggested that there is an optimum level of information which can inform better decision-making. Too little or too much information can evoke frustration and confusion, and is less likely to be perceived as useful (Schommer, Doucette, & Worley 2001).
Novelty of information
The novelty of information can affect how it is perceived and accepted. Information about technology which is new and/or unknown is more likely to be seen as high risk and therefore less likely to be accepted by the public (Slovic 2001).

Source of information
The source of information can also affect its acceptance. In relation to biotechnology, focus group participants rated doctors as the most trustworthy to provide balanced and honest information (MORI 1999). The least trusted sources of information included industry, retailers and the media, while governments were ranked as 15th out of 20 as the most trusted source.

Individualised risk information
A systematic review comparing individualised risk communication, based on for example family risk of a disease, age or epidemiological data, with general non-personalised risk information found that the former was associated with higher uptake of screening such as mammography and cholesterol testing (Edwards, Unigwe, Elwyn & Hood 2003). Tailored health communications have also been associated with increased physical activity (Bull, Kreuter & Scharff 1999), smoking cessation (Lancaster & Stead 2005) and increased fruit and vegetable intake (Oenema, Tan & Brug 2005).

Gain or loss-framed messages
The economic ‘prospect theory’ states that individuals are more likely to take a risk when confronted with information about loss (for example, “you may miss out on winning £100 if you don’t buy this lottery ticket”) and avoid risk when confronted with information about benefits (for example, “if you do not buy this lottery ticket you will have saved £1”). Similarly, the way health risk information is presented has been shown to influence health decision-making, for example, framing a message to emphasise loss (“40% chance of dying”) or gain (“60% chance of surviving”).

However, in terms of personal health behaviours, the interpretation of the loss- or gain-framed messages must be considered in context with the perception of the health behaviour (Rothman & Salovey 1997). ‘Detection behaviours’ such as
screening, could be seen as risky decisions as they might inform of illness in which case a loss-framed message would be more effective to promote the behaviour. For example, "if you do not attend screening a tumour could be left undetected and you may be less likely to survive" rather than a gain-framed message such as "screening detects tumours early; attendance gives you a greater chance of survival". Indeed, loss-framed messages have been found to increase engagement in breast self-examination, cholesterol screening and HIV testing (Rothman & Salovey 1997). However, some individuals may perceive screening to be a health-affirming behaviour and therefore would respond better to gain-framed information.

'Prevention behaviours' such as condom use could be considered desirable as they reduce the risk of future ill health. Prospect theory would predict that gain-framed messages would be effective in promoting them. Evidence was found to support this in both condom and sunscreen use. However, it is not known whether the perception of the behaviour as an effective preventative has a moderating effect (Rothman & Salovey 1997).

'Recuporative behaviours' (treatments or cures) could also be seen as beneficial and thus a gain-framed message would be more likely to be effective. Some evidence was found to support this but the studies only used hypothetical situations. In addition, it is not known whether the health decision was based on the perception of the efficacy of the behaviour or the salience of the desirable outcome (Rothman & Salovey 1997).

A more recent review of six studies undertaken in clinical settings suggested that uptake of screening practices is more likely when messages emphasise the disadvantages of not being screened (loss-framed) rather than the advantages of being screened (gain-framed) (Edwards et al 2001). However, the authors conclude that participants may not be as easily manipulated as it seems, as only two of the six studies revealed significant results.

Message framing may also interact with dispositional features of the individual. Research has shown that monitors and blunters can react differently to the way health risk information is presented. Monitors attend closely to health information
and retain greater and more accurate knowledge from it. They therefore tend to adhere better to recommended behaviours. However, monitors also tend to become more anxious. A loss-framed message may therefore increase their arousal to the point of disengagement from the health information. It is hypothesised that a neutrally-framed message is most effective in conveying health threat information to monitors.

On the other hand, blunters tend to distract from a health threat message rendering them less likely to retain accurate information. As a consequence they are less likely to become anxious and are less likely to adhere to recommended behaviours. Loss-framed messages therefore should increase the significance of the health threat thereby increasing the processing of information with consequent implementation of behavioural recommendations without increasing anxiety levels.

Miller et al (1999) tested these hypotheses by manipulating the framing of information given to patients about to undergo a cervical cancer diagnosis treatment (colposcopy) to be positively, negatively or neutrally framed. They also measured the following: participants’ attentional style using the Monitoring-Blunting Style Scale (MBSS: Miller, 1987); affective response by administering a scale measuring the impact of an event; a cognitive response of knowledge retention by asking true/false questions about the information they had received about the procedure; and, behavioural response by asking patients how often they had missed or cancelled previous appointments.

In addition to being characterised as monitors or blunters the MBSS can be used to distinguish between high and low scorers on each concept. Previous research has suggested that two separate scores may be more appropriate as monitor and blunter concepts are two separate dimensions (Myers & Derakshan 2000). In this case participants were classed as low or high monitors. As hypothesised, high monitors who received negatively framed information reported more intrusive thoughts than those who received neutrally framed information. However, this affective difference did not translate into differences in missed or cancelled appointments. Low monitors, as predicted, reported greater retention of information and less cancelling or rescheduling of appointments when the information was negatively framed than
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neutrally framed. However, negatively framed information did not result in the least degree of intrusive thoughts in these individuals. In other words, low monitors respond better to negatively framed information without experiencing emotional distress.

Research indicates that types of health risk information and the way it is presented can influence its interpretation and impact. The framing of genetic health risk information has not yet been investigated.

The following section focuses specifically on the impact of genetic information.

### 2.3 Impact of genetic health risk information

In addition to psychological factors affecting the interpretation of health messages, risk information in turn has psychological and behavioural effects. The increasing availability of gene-based health risk information has prompted research on the impact of such information.

**Behavioural impact**

The potential benefit of genetic tests complex disease risk factors is that test results may motivate people to take up healthier behaviours in order to reduce their risk of disease. However, research to date shows inconclusive support for this assumption.

Most early research in this area was conducted on the impact of knowledge of carrying the BRCA1/2 breast cancer genes or a strong family history of the disease on breast cancer screening uptake. A meta-analysis to determine the impact of risk assessment counselling on women with a family history of breast cancer reported that most studies showed improvements in screening behaviour (Meiser & Halliday 2002). An earlier review reported only modest increases in screening (Marteau & Lerman 2001) and other studies have shown differences in screening uptake according to age and level of education (Lerman, Croyle, Tercyak & Hamann 2002; Meiser & Halliday 2002).
Research on other diseases has been conducted, albeit to a lesser extent, which have failed to show positive impacts on health behaviours. Studies of individuals with bowel or prostate cancer or with a family history of heart disease found that they were no more likely to use screening or engage in risk-reducing behaviours respectively than the average risk person (Marteau & Lerman 2001). Moreover, although smokers who were told of a positive genetic test result showing an increased risk for smoking-related lung cancer reported increased intentions to quit this did not translate into actual quitting (Marteau & Lerman 2001).

Beliefs about the disease in question may affect the behavioural impact of the genetic test. Senior, Marteau, & Weinman (2000) suggested that people may interpret a genetic predisposition to an illness as uncontrollable which could actually deter people from engaging in risk reducing behaviours; they become fatalistic. However, results from a qualitative study did not support this. When individuals were told of an inherited risk for heart disease, they maintained their belief that the disease was controllable and adhered to dietary recommendations (Senior, Smith, Michie, & Marteau 2002).

**Emotional impact**

Much research has also been conducted on the emotional impact of genetic-based risk information. Although some studies have shown that individuals’ level of anxiety increases when faced with a positive genetic test result (Croyle et al 1997), this is not necessarily always the case. Other research found that anxiety may actually be reduced as uncertainty over genetic status has decreased and that some people may experience increased anxiety levels after negative test results possibly due to difficulties in adjusting to a different risk status (Wiggins et al 1992). Other evidence shows little change in levels of anxiety or depression after receiving negative test results, either in the short-term or long-term (Lerman et al 1996; Tibben, Timman, Bannick & Duivenvoorden 1997).

In 1999 these and other studies conducted so far and their respective data were reviewed and meta-analysed in order to provide a summary of research (Shaw, Abrams & Marteau 1999). Overall it was concluded that positive genetic results can cause increased short term anxiety, depression and distress although this does not
Chapter Two: Psychology and changing behaviour

persist in the long term. Moreover, there was no evidence to show adverse effects of negative results.

However, there may be some subgroups of individuals who are susceptible to adverse effects of predictive genetic testing. For example, Lerman et al (1998) found the highest levels of depressive symptoms in women who had undergone a breast cancer test in those who were highly stressed before the test. Other factors such as gender, social support, psychological coping resources, mood, existing perceived risk, pre-test distress and affected family members have all been shown to influence the emotional impact of genetic information, perhaps more so than the test result itself (Codori et al 1996; Lerman et al 2002; Marteau & Croyle 1998).

**Cognitive impact**

Concern has been raised that genetic risk information for complex diseases could be misunderstood (HGC 2002). Some research has been conducted on the interpretation and accurate recall of disease risk following genetic consultations. One study asked women about their perceived risk of breast cancer one week after attending a genetic consultation where they had been given accurate information on their family risk (Huiart et al 2002). Although in the majority of women the genetic consultation reduced inaccurate risk perceptions, a substantial minority inaccurately recalled their actual risk. Factors associated with incorrect recall of risk included inaccurate pre-consultation risk perception and feelings of anxiety or depression.

Other research has found that perceived risk is actually more accurate after genetic information. In a meta-analysis to determine the impact of genetic counselling on women with a family history of breast cancer, Meiser & Halliday (2002) found that the studies analysed showed significant, or trends towards, higher percentages of women accurately assessing their risk after counselling.

Research has also shown that beliefs about the causes of illness can determine perceptions of disease controllability (Marteau & Senior 1997). There may be a risk that genetic risk information causes individuals to become fatalistic – to believe that a disease is uncontrollable, with no hope of prevention or treatment. Results from a focus group study revealed that although the majority of participants rightly believed
that heart disease is caused by complex gene-environment-behaviour combinations
and that 'a gene for heart disease' means you have a heightened risk, a substantial
minority believed genes were the only risk factor and 'a gene for heart disease'
meant the disease was inevitable (Bates et al 2003).

Another aspect of fatalism can be seen in an experiment conducted by Wright,
Weinman & Marteau (2003). They designed a vignette study looking at the effect of
knowledge of a genetic predisposition to nicotine dependence on perceived control
of quitting and type of method of quitting preferred. Participants were told that
Zyban, a drug used to treat nicotine addiction, would be effective in those with a
positive test result but not in those with a negative result. They found that those in
the gene-positive group were more likely to choose Zyban and less likely to say they
would use willpower than those in the gene-negative group. The authors interpreted
this finding as individuals having "less faith in their own coping abilities" (p.229)
when they learn of a genetic predisposition to nicotine addiction.

Some would argue that this is evidence of fatalism – that gene-positive participants
resigned themselves to the fact that their nicotine addiction was not under personal
control, and that only drugs would cure it. However, it could be argued that learning
this information reduces one's self-efficacy for quitting; that quitting would be much
more difficult if the addiction is genetic. On the other hand, perhaps gene-positive
individuals choose the easier option (drugs over willpower) whereas gene-negative
individuals chose willpower because they had been told that Zyban would not work
for them.

There is also conflicting evidence of false reassurance – the perception that one's
risk is lower if a susceptibility gene variation is not found. The danger of this
perception is that unhealthy behaviours are continued, as individuals do not see
themselves as at risk. Evidence of false reassurance was found after a genetic colon
cancer susceptibility test (Lerman et al 1996) but not after a negative mammography
screening result (Drossaert, Boer, & Seydel 2001).

In a study to determine the impact of a genetic test for Alzheimer's Disease (AD) on
risk perception, adult children of people with AD were randomised to either the
intervention group where they were given a risk estimate based on age, gender, family history and the ApoE genotype, or the control group where their risk estimate was based on the same except for the ApoE genotype (Marteau, Roberts, LaRusse & Green 2005). Individuals who received a gene-negative result had a significantly lower perceived risk than the control group, although not to the extent that it could be classed as false reassurance.

Comparative optimism is the tendency to believe that we are more likely to experience positive events and less likely to experience negative events, compared to our peers. For example, Weinstein, Marcus & Moser (2005) reported that smokers tend to underestimate their relative risk, believing that they have a lower risk of developing lung cancer than the average smoker. This bias in risk perception may influence how individuals interpret their genetic test information. However, evidence for comparative optimism in the field of perceived risk of breast cancer is mixed. Some research has found that generally women have an optimistic bias about their risk of breast cancer (Katapodi, Lee, Facione & Dodd 2004). However, those women who perceived a higher risk were more likely to pursue genetic testing. In contrast, Gurmankin, Domchek, Stopfer et al (2005) found that before genetic counselling, women’s perceived risk of breast cancer was higher than their actual risk. Counselling resulted in lowering their perceived risk, but not enough to be accurate. There is evidence that unrealistic risk perceptions can be ‘debiased’ by emphasising the situation as severe and blameworthy (McKenna & Myers unpublished).

To date there have been few studies on the impact of genetic tests for complex disease risk factors. Of those that have (for example, Senior, Marteau & Weinman 2000), the information provided has been brief and may not have allowed the participant to fully interpret that information. This is unlikely to reflect how genetic information would be presented to an individual in real life. Even in the case of tests supplied directly to the public, with no health professional intervention, the interpretive information accompanying the test results is likely to be more detailed than that used in research studies so far.
Chapter Two: Psychology and changing behaviour

The following chapter draws together the issues discussed so far in Chapters One and Two to provide a rationale for the research described later in the thesis.
CHAPTER THREE

AIMS OF THE RESEARCH
3.1 Rationale for research

Genetic variations, in conjunction with environmental and behavioural factors, can influence the risk of developing complex diseases such as heart disease. It is well-known that health-related behaviours such as smoking, diet and physical activity contribute greatly to these prevalent and dangerous diseases of today. In turn, psychological factors affect the execution of such behaviours. The purpose of health risk information is to produce a desired outcome, such as the uptake of healthy behaviours. However, the way such information is presented may have subtle influences on this outcome.

Within twenty years the technology used to identify genetic variations which confer susceptibility to complex diseases is likely to be sufficiently developed so that it can help guide individuals in their efforts to maintain healthy lifestyles. At present there is mixed evidence to show that genetic test results are effective in motivating the uptake of healthy behaviours, although research to date could have imitated real life situations to a greater extent. If the knowledge about one’s genetic susceptibility to complex disease risk factors is shown to motivate people to become healthier it is important that this type of information is easily accessible.

Debates around the accessibility of genetic testing services to the public have discussed issues such as the confidentiality of genetic information, discrimination based on test results and genetic counselling. The current government agenda of promoting individual autonomy and responsibility for health suggests that genetic health risk information should be available in order for the public to use it to inform health-related decisions.

The research presented in this thesis describes the issues discussed during a public consultation exercise on the availability of genetic testing services to the public. It also discusses how useful the consultation was in terms of representation of views and the potential impact of regulatory decisions on the principle of increasing individuals’ responsibility for actively maintaining their health. The thesis also
Chapter Three - Aims of the Research

presents results of an experiment investigating the potential impact of genetic information, and the psychological factors which may influence this impact.

3.2 Stakeholder analysis

Aims
The aims of this study were to explore the views of stakeholder group representatives on genetic tests for complex disease risks, in particular the regulation of public access to genetic testing and public consultation relating to this. A further aim was to investigate potential biases in public consultations.

Research questions:

1. To what extent did stakeholders believe the public consultation to be comprehensive and objective?
2. Do stakeholders agree with the recommendations made by the HGC?
3. What issues do stakeholders believe to be the most important and relevant to complex disease risk genetic testing and its access-related regulation?
4. Do stakeholders’ views on individual autonomy over health information differ when acting as representatives of their organisation than those they hold as individuals?
5. Do stakeholders’ views on individual responsibility for health differ when acting representatives of their organisation than those they hold as individuals?

Objectives
In order to answer the research questions, the following objectives will be achieved:

- Develop and pilot an appropriate semi-structured interview schedule;
- Explore views and opinions of individuals listed as responding to the public consultation, or known to have knowledge and/or interest in the area by conducting and qualitatively analysing interviews;
- Critically evaluate the conduct of the HGC-led consultation; and,
Chapter Three - Aims of the Research

- Consider the impact of access-related regulatory policies on genetic tests for complex disease risk information.

3.3 Experimental questionnaire

Aims

The aim of this study was to investigate the potential value of genetic tests for complex disease risk factors in addition to individuals’ concerns about the use of genetic information.

Research questions:

1. What is the impact of genetic health risk information on anxiety, perceived risk of heart disease and intention to change diet?
2. Do demographic factors or health indicators predict reactions to the test results?
3. To what extent can psychological variables and beliefs predict the impact of genetic health risk information?
4. What influence does information presentation have on the impact of genetic health risk information?
5. To what extent are individuals concerned about the use of genetic health risk information?
6. To what extent did individuals understand the genetic and nutritional information given to them?

Objectives

In order to answer the research questions, the following objectives will be achieved:

- Evaluate existing evidence relating to the impact of genetically-based health risk information;
- Evaluate evidence relating to health behaviour change;
- Develop, pilot and conduct an experimental vignette-style questionnaire on a non-clinical population presenting hypothetical genetic test results and measuring emotional, cognitive and behavioural outcomes; and,
Chapter Three - Aims of the Research

- Critically evaluate the potential value of gene-based complex disease risk information.
CHAPTER FOUR

RESEARCH DESIGN AND METHODOLOGY
4.1 Qualitative research

Qualitative research methods describe observable phenomena in words rather than numbers. This method of research is often used to explore a little known or complex subject area or to allow a more in-depth exploration of a topic. It is often used to generate hypotheses for future testing, although it is appropriate to consider possible explanations for the findings.

4.1.1 Sampling strategies

Four main sampling methods are used in qualitative research (Bowling 2002; Smith 2002). Convenience sampling describes the recruitment of participants for reasons of convenience in terms of location, ease of recruitment or likelihood of participating. Although this may save on resources and is acceptable in some cases it is likely to be unrepresentative of the target population. A technique called ‘snowballing’ is sometimes used to recruit unknown participants where preliminary respondents are asked to suggest others who may be eligible to take part in the research. Purposive samples involve participants who are deliberately and non-randomly recruited for specific reasons. For example, in order to understand a particular group of people or event, or to gain the most informed opinions (eg. Abraham & Lewis 1999).

Purposive sampling can also be used to maximise variability in a sample, for example, recruiting participants on the basis of age, employment status or GP practice and so on. Representative sampling involves recruiting a specific number of participants, for example, from all pharmacies in one borough, with the aim of ensuring some degree of representativeness in the sample. Finally, theoretical sampling involves the recruitment of participants until no new insights into the target research area are gained. This is often referred to as reaching ‘theoretical saturation’. This type of sampling requires coding and analysis of data being carried out concurrently with the sampling process.

Sample sizes are often small in order to allow the subject area to be explored and analysed to such detail not possible with larger numbers. The time consuming and complex nature of the data collection and analysis also necessitates small samples. However, due to this, representativeness of the sample is therefore often a criticism.
Findings from one sample cannot always be generalised to other individuals, situations or points in time, although speculations, rather than generalisations, can be made to similar situations provided evidence and rationale is given (Borman, LeCompte & Goetz 1986).

4.1.2 Qualitative research methods

Observation
Observational techniques are used to understand behaviours, actions, activities and interactions (Bowling 2002). It is based on the need to understand a particular group, their traditions, beliefs, rituals and experiences. The researcher can be participative or non-participative within the target group. As a participant, the researcher may get to know the group well and gain their trust and honesty. On the other hand, a non-participant researcher can be sure to observe the group without influencing their behaviour by their presence.

Interviewing
Interviewing techniques are used to gain an in-depth understanding of people's knowledge, attitudes, experiences and beliefs. Interviewees provide information in their own words with prompting from the interviewer. The interview can vary in its degree of structure. Unstructured interviews may have a checklist of topics for guidance but the interview essentially takes an unstructured course. Semi-structured interviews may have more specific guidance topics and/or some open ended questions but again, the interview takes a unique course. If the research is following a grounded theory approach the researcher may want to explore potential theories which have been developed from previous interviews and thus the guidance topics may change from interview to interview.

An essential characteristic of the researcher is to be flexible and receptive (Smith 2002). Each interview will be different and topics will be discussed in varying orders and depths, depending on the interviewee. The researcher must be prepared to investigate new areas, to encourage the interviewee to expand on some subjects yet direct the focus of the interview towards the research agenda if it deviates.
Focus groups
Focus groups are a group interviewing technique with the aim of exploring views and opinions of several people at one time, as opposed to one-to-one interviews. The other key feature of focus groups is the interaction between participants. This tends to encourage the generation and discussion of a wider range of ideas and issues than would arise in individual interviews (Smith 2002). However, the involvement of several participants may preclude deeper examination of individual’s issues. In addition, some individuals may be more involved in the discussions than others.

4.1.3 Qualitative data analysis
There are numerous ways of analysing qualitative research and each is useful for a particular purpose, or for a particular level of analysis. Table 4.1 presents descriptions of some of the methods, and their suitability as a technique for this phase of the project (Bowling 2002, Smith 2002).
## Table 4.1: Types of qualitative analysis

<table>
<thead>
<tr>
<th>Method</th>
<th>Description</th>
<th>Suitability for this study</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Content analysis</strong></td>
<td>A review of documents, newspapers, reports or narratives. Occurrences of words or themes are counted then subjected to statistical analysis to note how often or where particular citings occur.</td>
<td>Not appropriate as: 1) It required <em>a priori</em> knowledge. It was important in this study, the data was analysed with as few preconceptions as possible. 2) A deeper level of analysis was required to describe stakeholders’ explanations and reasons for views and beliefs.</td>
</tr>
<tr>
<td><strong>Discourse analysis</strong></td>
<td>Attempts to uncover and explain the structure of communication. Natural dialogue itself is the focus of the investigation - examining the way questions, answers and statements are made or studying accepted ‘rules’ of dialogue such as not interrupting.</td>
<td>Not appropriate as: 1) The detailed structure of conversation was not a research aim of this study.</td>
</tr>
<tr>
<td><strong>Grounded Theory</strong></td>
<td>As described above, Grounded Theory aims to generate, develop and verify theory from the data. Data is analysed concurrently with data collection.</td>
<td>Not appropriate as: 1) The study was exploratory with the aim of describing views and opinions rather than generating theory.</td>
</tr>
<tr>
<td><strong>Interpretative Phenomenological Analysis (IPA)</strong></td>
<td>IPA was developed specifically within the discipline of psychology. It is phenomenological in that it is concerned with an individuals’ perception of events and experiences, and is interpretative in that it acknowledges the researcher’s role in making sense of the individual’s experiences.</td>
<td>Although this approach would have been suitable for this study, it was decided that the charting stage of framework analysis (see below) would be more useful in comparing within and between stakeholder groups.</td>
</tr>
<tr>
<td><strong>Framework Analysis</strong></td>
<td>This method was developed in the context of applied policy research. The analysis is grounded in the original data but also takes into account <em>a priori</em> ideas. The data is analysed systematically and repeatedly and is presented in a way which enables comparison between and within cases and allows descriptions and explanations to be easily tracked and judged.</td>
<td>This was deemed the most appropriate method to use for this study for the reasons described in Section 5.1.3.1 below.</td>
</tr>
</tbody>
</table>
Chapter Four — Research design and methodology

4.1.3.1 Framework analysis

This method was developed in the context of applied policy research by a UK qualitative research unit within now known as the National Centre for Social Research. The method is generic and its principles versatile so that data from any discipline can be used. It has therefore been widely applied in areas such as self-harm, influences on hospital admission, cardiac rehabilitation services and attitudes to the deregulation of emergency contraception (Chew-Graham et al 2002; Griffiths et al 2001; Tod, Lacey & McNeill 2002; Harper & Barratt 1998).

There are five stages involved in framework analysis:

*Familiarisation:* In this stage the researcher gains an overview of the data by listening to recorded interviews and re-reading transcripts. Ideas and recurrent themes and issues are noted down to be explored later.

*Identifying a thematic framework:* According to Ritchie and Spencer (1994) the thematic framework is constructed from a priori issues (such as those which structured the research questions), themes that emerge from the respondents themselves and those which have been interpreted by the researcher. The first version may be largely based on a priori themes but as the framework is applied to transcripts, a more detailed index may emerge. Categories are refined as the experiences and attitudes of the respondents are conceptualised.

As well as serving to sort the data into key issues and themes, the framework, or index, also allows the data to be displayed and managed in such a way as to make retrieval and cross-referencing simple (Table 5.3 in Chapter Five).

*Indexing (coding):* In this stage the framework is applied to the text. The data are annotated according to the framework (example in Appendix). It may sometimes occur that a section of text relates to more than one theme; in this case, both or all references are noted against the text. The researcher can then see patterns emerge. One of the criticisms of qualitative analysis is that it is subjective and open to
interpretative bias. By indexing the text in this way, it provides transparency of the process which others can view and judge.

**Charting**: In this stage the data is gathered together. Data can be arranged in a matrix or diagram either by theme across all respondents or by respondent across all themes in order to aid data interpretation. The annotated sections of text are summarised and entered into the chart, with reference to the original text.

**Mapping and interpretation**: Here the researcher begins interpretation and explanation by comparing individuals and themes, and locating patterns and associations. This may be done in a variety of ways depending on the aim of the research and emergent themes.

Framework analysis was deemed the most suitable for the purposes of this study for several reasons. Firstly, this method acknowledges that the emerging themes will originate from both the researcher and the respondent. Although this study was exploratory, there were specific areas which warranted discussion.

Secondly, the method allows for the description of data by grouping into themes which is an effective way to describe, compare and present the data. In addition, by coding portions of data in this way and referencing back to the original document, the retrieval of data is simplified. One can see where a particular theme has occurred across all documents, or can easily be referred to a portion of dialogue which can be examined in greater detail. Related to this, the fourth reason for using this method is that the way the information is presented in the charts allows for comparison within and between cases. The intention of the research was to examine individual views, as well as compare differences and similarities between individuals in particular ‘stakeholder groups’, for example those in ‘industry’, and between individuals across all groups.

The method also allows the analytic process to be transparent. This makes it easy to trace interpretations by the way the text is referenced and charted throughout. It also demonstrates to others the analytic process and organisation which serves to ensure the credibility of the findings. The method was also suitable because the qualitative
software package QSR NVivo v1.3 is built in such a way to facilitate and complement this type of analysis. For example, it allows you to assign codes to sections of data, and to look within or across documents for occurrence of codes. NVivo is a more sophisticated programme than others which are commonly used for qualitative analysis. For example, ATLAS.ti is not compatible with Windows software and the searching facilities are limited. NVivo's predecessor NUD*IST also has some disadvantages such as it uses fixed 'text units' when highlighting to code text and has a less flexible memoing facility, only allowing one per node or document.

4.1.4 Ensuring credibility in qualitative research

Qualitative analysis is, by nature, subjective, interpretative and exposed to bias from the researcher who inevitably brings preconceived ideas to the data (Borman, LeCompte & Goetz 1986). Generally, one researcher conducts both the research and the analysis and there is a risk that personal views can influence the conclusions drawn. However, some have argued that quantitative and statistical methods are developed and chosen, and findings interpreted by a researcher who may also have predetermined ideas (Patton 1980). Patton argues that absolute objectivity is impossible to attain and would ignore the intrinsically social and human nature of research. Nevertheless, there are measures that can be taken to reduce this risk and enhance the quality and credibility of the findings.

Researchers should be aware of their potential biases and actively search for them and challenge their analyses. Different researchers will inevitably interpret the meaning of the data differently. To counteract this interpretation bias, data can be analysed by others to ensure consistency of coding and conclusions. In addition, triangulated methods can be used to ensure reliability of results. This involves using data gathered from other researchers, sources (such as published literature) or time points for comparison. Transparency of the analytic process is also important to enable the reader to trace explanations and conclusions by the way data is recorded, referenced and charted.
4.2 Quantitative research

Quantitative research explores and quantifies relationships, frequencies and characteristics by using numerical data and statistical analysis to summarise the findings. Its advantages are that a lot of data can be obtained in a relatively short period of time and at relatively low costs. The aim of quantitative research is to gather information from a sample of sufficient size in order to be able to generalise the findings to the wider population (Smith 2002).

4.2.1 Quantitative study design

Surveys and questionnaires are the most widely used tools for gathering quantitative data in social science research and using previously validated instruments allows for the comparison of results across studies using that particular measure. In addition, studies using the same questionnaire can provide information on its validity in different populations and settings. However, the validity of an instrument used in novel conditions will be uncertain.

There are several ways a questionnaire study can be designed however. A questionnaire can be used in a cross-sectional study where participants provide data at one time point. Within this cross-sectional design, the researcher may want to compare more than one group of individuals. The groups may already be distinct from one another (e.g. by gender, from different countries, etc) or they could be randomly assigned to different groups at the beginning of the study (an experimental approach). For example, participants would be exposed to different conditions and then asked to respond to the questionnaire. Responses from individuals in different conditions would then be compared.

Cross-sectional studies typically investigate current and/or past behaviour or attitudes. Retrospective studies though can be prone to biases in recalling data. Another disadvantage of the cross-sectional design is that cause and effect relationships cannot be established. Longitudinal studies however can provide this information. Participants are measured at several time points and differences over time are compared. However, this design also has disadvantages in that it can be
more resource costly and participants may drop out of the study or become untraceable between time points.

Although qualitative and quantitative methods have their own unique qualities, they can also complement each other well in an investigation to provide balanced and thorough data. Qualitative research can take place initially to gain preliminary information to inform the development of a quantitative instrument or to provide in-depth detail and to stimulate theories and hypotheses which can be tested or validated with quantitative methods. In addition, quantitative research can be used initially to highlight topics to be pursued in depth by qualitative methods or a subsequent qualitative study may help explain findings gathered from a quantitative study. These methods of triangulation aim to draw on the strengths of each method in order to produce robust findings.

4.2.1.1 The vignette technique

Vignettes are stories about individuals or events which are used in social sciences research to elicit beliefs, attitudes and perceptions. They are often based on real-life case studies and are examples of situations, or are hypothetical situations, designed to reflect events in the real world to which participants are asked to respond with what they would do in that situation, or how they think a third person might respond (Hughes 1998).

They are usually used when the equivalent real-life situation is not ethically or feasibly possible. A search on PsychInfo for studies published in 2004 showed research topics using a vignette design included suicide, drug injecting, domestic violence, and attitudes towards homosexuality. Genetics is another area where the vignette technique is used. In a study by Senior, Marteau & Weinman (2000) participants were asked to imagine receiving genetic test results which indicated the risk for a disease and then asked to respond to a series of questions. Other areas of research have included genetic counselling and reproductive decision making.

One criticism of the technique is that people may respond differently to real life situations than those given in the vignette. This may be because the vignette does not provide enough information for the individual to make a decision or that it neglects
Chapter Four – Research design and methodology

the social and environmental factors which affect decision-making in real life. Additionally, individuals may respond as they should rather than as they would, or as how other people would react rather than themselves or vice versa (Hughes 1998). However, it could be argued that these criticisms could apply to other research instruments.

4.2.1.2 Reliability and validity of survey instruments

Validity refers to the extent to which an instrument actually measures what it purports to measure. New instruments can be subjected to several tests to ensure validity. Reliability refers to the extent that an instrument can elicit the same responses over time and that items within the instrument are measuring the same phenomenon. Questions should be unambiguous and clear in order to avoid confusion, poor response rates or misleading responses. Piloting of an instrument should uncover these kinds of issues.

4.2.2 Samples

There are various ways of identifying potential participants for a quantitative study. A random sample, where every member of the population has an equal chance of being included in the study, maximises the chances of the sample being representative of the whole population, and therefore the results can be generalised. In order to use this sampling technique the researcher must have access to the whole population in order to select from it randomly, often using random number selection, or every n\textsuperscript{th} person.

Cluster sampling involves the random selection of clusters within a population (for example, randomly selecting UK counties) and subsequent random selection of individuals within those selected clusters. If a researcher wishes to compare two or more populations he may wish to use stratified sampling. Random samples are taken from each population in order to ensure equal numbers in each group.

Convenience sampling for quantitative studies has the same principles as those for qualitative research. The main criticism of this technique is the uncertainty one has of the representativeness of the sample. ‘Volunteer’ studies are open to the same
criticism as volunteers may have different characteristics to non-volunteers which presents a self-selection bias. However, these characteristics may not be related to the variables under investigation and therefore the generalisability of the findings would not be compromised (Smith 2002). Similarly, individuals who agree to participate but for some reason do not provide all the data or who drop out at some stage may differ from those who fully complete the study. Ideally, the characteristics of responders and non-responders would be compared to identify any differences, although often this is not possible.

Achieving a high response rate increases the confidence with which you can generalise the findings to other members of the population. There are several ways to maximise response rate. Reminder letters or telephone calls or repeat mailings of the questionnaire can be sent. These can be targeted at non-responders if they can be identified. Distributing questionnaires by email has also been shown to be quicker, more convenient and yield more responses (Shannon & Bradshaw 2002). Some researchers use incentives to encourage responses. Monetary incentives have been shown to double response rates (Edwards et al 2002).

Sample sizes need to be sufficient to allow differences between groups to be detected, that is, have sufficient statistical power. A power calculation can be carried out which estimates the sample size according to levels of significance and desired power level. The smaller the sample, the more risk there is of a Type 2 error. That is, erroneously accepting the null hypothesis (and therefore rejecting the alternative hypothesis that there are differences between groups). In contrast, an overly large sample size may risk a Type 1 error of erroneously rejecting the null hypothesis as statistically significant results are more likely to be found in larger samples.

4.2.3 Data analysis

Data cleansing

Inputting large amounts of data into a computer spreadsheet or statistical package programme can be repetitive and mistakes are easily made. Checking the entered data for accuracy is essential. A common way of cleansing the data is to take a
percentage of the sample and compare the entered data with the raw data from the questionnaires. This calculation is used to generate an error rate:

\[
\text{Error rate} = \frac{\text{number of errors}}{(5\% \text{ of sample}) \times (\text{number of rows} \times \text{number of columns of database})} \times 100
\]

**Data analysis**

Depending on the questionnaire design and type of data there are various statistical analyses which can be conducted on the sample in order to investigate differences between groups or over time, whether one variable predicts the occurrence of another or whether some variables are associated with each other. One of the main considerations when choosing a statistical test is whether the data meets the criteria for a parametric test. Parametric tests are more precise and powerful since the data used is of 'higher' quality. In order to use a parametric test data must be of at least interval level, that is, be continuous and have equal intervals regardless of location on the scale. An example of interval data is temperature. Ratio level data can also be used which has all the criteria of interval data and, in addition, has a true zero which indicates an absence of the measured characteristic. An example of ratio level data is weight or age. Additionally, data must be normally distributed and have equal variances within the populations studied (homogeneity of variance) in order to be subjected to parametric analysis. Table 4.2 presents a brief description of statistical analyses, the type of data required to conduct it and refers to examples in Chapters Five and Six.
### Table 4.2: Types of statistical analyses

<table>
<thead>
<tr>
<th>Comparing two different (unrelated) groups</th>
<th>Parametric</th>
<th>Non-parametric</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>UNPAIRED T-TEST</td>
<td>MANN-WHITNEY TEST</td>
</tr>
<tr>
<td></td>
<td>Eg. Anxiety levels in Gene and No Gene groups (see Section 6.3.3)</td>
<td>Eg. Intention to change diet in Gene and No Gene groups (see Section 6.3.3)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Comparing the same group at two different time points</th>
<th>PAIRED T-TEST</th>
<th>WILCOXON TEST</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eg. Anxiety levels before and after test results (see Section 6.3.3)</td>
<td>Eg. Opinions as a representative of an organisation and as an individual (see Section 5.3.3)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Comparing at least three unrelated groups</th>
<th>ONE-WAY ANALYSIS OF VARIANCE (ANOVA)</th>
<th>KRUSKAL-WALLIS TEST</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eg. Anxiety levels in different age groups (see Section 6.3.3)</td>
<td>Eg. Intention to change diet in different age groups (see Section 6.3.3)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Comparing at least three groups at different time points</th>
<th>REPEATED MEASURES ANOVA</th>
<th>FRIEDMAN TEST</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Investigating the extent of the relationship between two continuous variables</th>
<th>PEARSON'S CORRELATION</th>
<th>SPEARMAN'S CORRELATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eg. Investigating the association between activity levels and anxiety levels (see Section 6.3.3)</td>
<td>Eg. Investigating the relationship between self-rated health and intention to change diet (Section 6.3.3)</td>
<td></td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th>Predicting one value from another measured variable</th>
<th>SIMPLE (NON)LINEAR REGRESSION</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Predict one value from several measured variables</th>
<th>MULTIPLE (NON)LINEAR REGRESSION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eg. Predicting anxiety levels from several psychological variables (see Section 6.3.4)</td>
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</tbody>
</table>

### Reliability analysis of scales

In order to test for an instruments' internal reliability, that is, that all items within the instrument are measuring the same phenomenon, the Cronbach's alpha method is
often used. Cronbach’s alpha is a statistic between 0 and 1 which reflects the correlation between items. A figure of above 0.7 is generally considered to be acceptable (Smith 2002). The distribution of scores should also be checked for normality. If scores are non-normally distributed, non-parametric tests should be used when conducting statistical analysis with that measure.
CHAPTER FIVE – STAKEHOLDER ANALYSIS:

Method, Results & Discussion
5.1 Introduction

The aims of this study were to explore the views of stakeholder group representatives on genetic tests for complex disease risks, in particular the regulation of public access to genetic testing and public consultation relating to this. A further aim was to investigate potential biases in public consultations. This chapter explains the methods used to undertake the stakeholder interviews and fully describes the analysis and results. It discusses the findings in the context of other research and considers its implications. Limitations of the study are presented and areas for future investigation are proposed.

5.2 Method

For the purposes of this particular subject area, which had not been investigated previously, it was appropriate to use a mainly qualitative design in order to explore the area and allow for spontaneous discoveries. By using quantitative methods one could only report on what had been measured. Important factors that had not, or could not, be measured would have been omitted.

5.2.1 Piloting the interview schedule

An interview schedule was designed and piloted. It consisted of several semi-structured questions based on issues which had already been raised during the HGC consultation. In addition some questions were designed to elicit the capacity of public consultations to accurately represent the views of the public and representative groups.

The interview also included some supplementary quantitative measures. These included a checklist of issues and concerns, again based on those discussed during the HGC consultation, which participants were asked to rate in terms of importance when deciding on regulatory policy. Two visual analogue scales were included on which participants were asked to indicate the balance of their opinion on the relative desirability of protecting the public through controlling access to genetic tests, as opposed to promoting greater individual responsibility via free access to such tests (see Figure 5.1). Participants were asked to respond as representatives of their
organisations, and then again with their private views. The scale had ten points, although this was not explicitly indicated.

Figure 5.1: Individual and organisational views on access and responsibility for health

| Individuals have a right to free access to information about their genetically determined health risks. Tests should be available to buy freely as well as being available through the NHS | The public needs protection from exploitation and needless anxiety. Tests should only be available through the medical profession |
| Adults are responsible for their own health. We need to move from paternalism to individual choice and autonomy | People at risk of illness are often the most vulnerable. They need professional direction. |

Additionally, participants were asked to show their agreement on a five-point Likert scale to twelve statements relating to public access to genetic information.

Although using quantitative measures within a qualitative interview is unusual, they offered an additional dimension to the research. In addition, they provided a level of triangulation where responses to, say, the checklist of concerns could be compared against the discussion of concerns raised during the interview. Although it is acknowledged that using statistical analysis is not common on small samples, the results give an indication of trends which merit discussion and could be pursued in future studies.

The instrument was piloted amongst four individuals who had knowledge of genetic tests and related issues, and who would have been eligible and appropriate to participate in the study. The instrument was judged viable and, following minor amendments, was used for the rest of the study.
5.2.2 Participants

Stakeholders are individuals, organisations or bodies who have a share or an interest in an activity. For the purposes of this study they were defined as those who were listed as responders to the HGC consultation document (Department of Health, 2003a), or who were known to have an interest in the outcome of the debate around access to genetic testing. For example, cancer charities who represent people affected by cancer, a disease whose risk factors could be identified by genetic tests, and professional bodies representing pharmacists, who may have considerable involvement in the supply of genetic tests in the future.

Individuals from five principal groups, based on categories created by the HGC, were contacted (see Table 5.1). This was a purposive, non-random sample; the individuals were contacted because of their known interest in or knowledge of the area. Responses therefore reflect informed and illustrative rather than random opinions. This method of sampling is accepted and has been used previously in research concerning health care regulation (Abraham & Lewis 1999). Although it cannot be said that these individuals held views which were representative of their stakeholder group, the sample as a whole reflected the range of interested parties who took part in the HGC consultation.

Table 5.1: Organisations and individuals represented in interviews.

<table>
<thead>
<tr>
<th>ACADEMIC</th>
<th>POLITICAL/REGULATORY BODIES</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Consultant in public health medicine</td>
<td>• Labour MP</td>
</tr>
<tr>
<td>• Professor of cardiovascular genetics</td>
<td>• Liberal Democrat MP</td>
</tr>
<tr>
<td>• Professor of cancer genetics</td>
<td>• Medicines and Healthcare products Regulatory Agency</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>INDUSTRY</th>
<th>PROFESSIONAL BODIES</th>
</tr>
</thead>
<tbody>
<tr>
<td>• British In-Vitro Diagnostics Association</td>
<td>• British Association of Nutritional Therapists</td>
</tr>
<tr>
<td>• Proprietary Association of Great Britain</td>
<td>• Royal Pharmaceutical Society of Great Britain</td>
</tr>
<tr>
<td>• Sciona Ltd</td>
<td>• British Medical Association</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PATIENT/CONSUMER GROUPS</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>• Consumers’ Association</td>
<td></td>
</tr>
<tr>
<td>• Genetics Interest Group</td>
<td></td>
</tr>
<tr>
<td>• CancerBACUP</td>
<td></td>
</tr>
</tbody>
</table>
5.2.3 Recruitment

Participants were approached by email, telephone or letter and invited to take part in the study. In order to investigate as many differing views as possible, breadth of representation was aimed for, rather than depth. Participants were therefore recruited until the sample was sufficient to allow a broad range of views to be elicited - at least three individuals from each stakeholder group. Eight individuals declined to participate (two did not reply, five felt that they were not informed enough to participate, and one did not have an official position on the subject). A further six were willing to take part but were unable to do so due to time constraints. A total of sixteen semi-structured interviews were therefore conducted.

5.2.4 Conducting the interviews

All interviews were recorded by minidisk with the participant's permission. Assurance that ethics committee approval for the study was not required was obtained via the supervisor. All participants signed a form requesting consent and assuring confidentiality.

Interviews were conducted in June and July 2003. Other than in one case, interviews were conducted in the interviewees' workplaces. In the exceptional instance the interview was held at the University of London School of Pharmacy. Although all topics were discussed, the interviews differed in terms of order and duration. They lasted for between 30 and 75 minutes.

5.2.5 The analysis

Interviews were transcribed verbatim, read thoroughly and text was coded into 'nodes', or themes using the software package QSR NVivo v1.3.

Credibility of the analysis was ensured in several ways. Firstly, a second researcher checked the text attributed to themes and confirmed its appropriateness. Secondly, the themes were scrutinised to ensure that the text attributed to each was suitable, and new themes were looked for which may not have emerged during the first round of analysis. New codes were constructed and the process of examination and verification was repeated. This iterative process of coding, analysing and verifying
ensures that all data has been closely examined several times and that underlying messages have been identified, further ensuring credibility.

Thirdly, original text was annotated according to the framework of emergent themes for both data management purposes, and to provide a transparency of the analysis. Transparency can allow the analysis to be scrutinised by other researchers to ensure credibility. Data was then organised into a matrix of themes by stakeholder group. This aided data interpretation and allowed patterns, associations and comparisons to be seen and made more clearly.

The Kruskal-Wallis non-parametric ranking test for independent groups was performed on the responses to the checklist of important issues to identify how each of the stakeholder groups ranked them. This was then compared with interview data.

The analogue scale consisted of a line with 10 unmarked notches. By measuring the distance of the mark from the left, a quantitative value of between 0 and 10 was obtained. Median values for these scores were calculated. A Wilcoxon Signed Ranks test for repeated groups was performed to see whether personal views were significantly different to views given as representative of the organisation. The sum of the visual analogue scale scores (individual views) was also explored and low or high scorers identified. Interview data for low and high scorers was then explored.

Frequency counts were calculated for the Likert-style responses to twelve statements. In addition, Spearman’s rho non-parametric correlations were performed to identify possible associated views.

It is important to note that using statistical analysis on such a small sample is not usually recommended, as it is unlikely to be representative of a larger population. However, statistics are being used here not to generalise to a wider population, but rather as an additional exploratory tool for general analysis. Results are presented in this thesis that, despite the small sample size, reached significance, or indicated trends which would be interesting to pursue further.
5.3 Results and discussion

This section reports the analysis of the stakeholder interviews and considers the findings in the context of relevant research and policy developments in addition to discussing possible interpretations of interviewees' views and opinions. Stakeholder group status refers only to that assigned for the purposes of this study. Although interviewees' views are likely to be indicative of those of the stakeholder group as a whole it is not assumed that they are representative of the latter.

5.3.1 Emergent codes and themes

Due to the nature of the interview not all questions were explicitly asked (for example, if participants had already discussed the topic) and not all questions were asked in the same order.

In the same way, some codes emerged as a result of particular questions asked, for example, perceptions of the HGC consultation and attitudes towards HGC recommendations. Other codes, however, emerged from participants' free discussion of the area. Table 5.2 shows the codes and their descriptions.
### Table 5.2: Primary code description and interpretation

<table>
<thead>
<tr>
<th>Code Description</th>
<th>General findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.1 Perception of HGC consultation</td>
<td>Perception of the HGC consultation on direct public access to genetic testing services. There were varying levels of familiarity with the conduct of the consultation and mixed views on how fully representative it had been, particularly regarding the public views.</td>
</tr>
<tr>
<td>1.2 Attitude towards HGC recommendations</td>
<td>Views on the recommendations made by the HGC based on the consultation. There was mixed familiarity with the recommendations made by the HGC. There was a general consensus that they were appropriate although some felt that they should have been implemented immediately.</td>
</tr>
<tr>
<td>2.0 Perception of industry sector</td>
<td>Stakeholders’ perception of the genetic testing industry. Some participants seemed to have a general sense of mistrust towards the industry in terms of how DNA samples are handled and stored, the accuracy of information they provide and their interest in selling genetic tests.</td>
</tr>
<tr>
<td>3.0 Genetic test providers</td>
<td>Comments on potential suppliers of genetic tests. Opinions were varied on who was best placed to provide genetic tests; some participants favoured pharmacists and some favoured GPs. Adequate training and knowledge was held to be a mandatory prerequisite.</td>
</tr>
<tr>
<td>4.0 Consent</td>
<td>Comments on consent to genetic tests. This issue was not raised by many participants in free discussion. It was generally mentioned in relation to children, as a group who was unable to give informed consent. Participants generally felt that it was a non-issue as parents would only use a genetic test in the best interests of the child.</td>
</tr>
<tr>
<td>5.0 Confidentiality</td>
<td>Concerns about the confidentiality of genetic information once a test has been conducted. Questions included: What happens to the information? How is it stored? Is it used again? Who has access to it? Most participants felt strongly about this issue and although the general opinion was consistent, there were subtle differences in reasoning. Some participants were concerned about use of genetic data by police and some were concerned about how private companies would keep data confidential. Others believed that data kept on NHS records would be the least confidential.</td>
</tr>
</tbody>
</table>


<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
<th>General findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>6.0</td>
<td>Discrimination</td>
<td>Attitudes towards the potential of discrimination on the grounds of one’s genetic information. Most participants had a firm view on discrimination. All stakeholder groups but one felt that most genetic data would not be predictive enough for discrimination.</td>
</tr>
<tr>
<td>7.0</td>
<td>Genetic ‘exceptionalism’</td>
<td>Views on the possible uniqueness of genetic data as opposed to other health information such as family history. Most participants seemed well informed enough to hold firm views on this issue. All stakeholder groups felt that genetic information was no different to other medical information used at present.</td>
</tr>
<tr>
<td>8.0</td>
<td>Quality of genetic tests</td>
<td>Comments on the validity, reliability and utility of genetic tests. A few participants felt that it was important for the government to ensure the accuracy, reliability and validity of genetic tests.</td>
</tr>
<tr>
<td>9.0</td>
<td>Validity and usefulness of complex disease risk assessment</td>
<td>Opinions on the validity and usefulness of genetic tests for complex disease risk factors. Most participants had opinions on this issue, many of them strongly held. Some groups felt that complex disease risk tests had limited usefulness yet some felt that they would be useful in motivating people to prevent illness.</td>
</tr>
<tr>
<td>10.1</td>
<td>Lay perception of genetics</td>
<td>Participants’ beliefs about the lay perception of genetics, and what influences this perception. Several participants held similar views about this issue; that is, there is a lack of understanding amongst the public about genetics and that the public’s perception of genetics is deterministic and negative.</td>
</tr>
<tr>
<td>10.2</td>
<td>Public understanding of information</td>
<td>Participants’ beliefs about the lay understanding of genetics and genetic information. Many participants had firm views which were similar across all stakeholder groups; that is, that the public generally misunderstand genetic information, particularly relative risk.</td>
</tr>
<tr>
<td>10.3</td>
<td>Impact of knowledge of genetics</td>
<td>Beliefs about the impact of personal genetic information on individuals and their families. Most participants held strong views about the impact of genetic information. Different views were expressed within and between stakeholder groups in relation to emotional and behavioural impact.</td>
</tr>
<tr>
<td>10.4</td>
<td>Information, support and counselling</td>
<td>Comments on the information, support and counselling that may accompany genetic tests and test results. Most participants felt strongly that the availability of information, support and counselling is essential.</td>
</tr>
<tr>
<td>11.0</td>
<td>Need for public education</td>
<td>Comments on the need for public education on genetics. Few participants mentioned this issue; those who did believed that public education was necessary to dispel misconceptions about genetics and allow tests to be used appropriately.</td>
</tr>
</tbody>
</table>
Table 5.3 displays findings by code and stakeholder group. References to the original text are also given (see Appendices page 263 for an example of an annotated interview transcript). The reference given represents the participant (eg AC1), and the section and paragraph of the transcript which refers to the code under discussion (eg. S3: P2).
Table 5.3: Interview data by code and stakeholder group

<table>
<thead>
<tr>
<th></th>
<th>Academic</th>
<th>Political/ regulatory body</th>
<th>Industry</th>
<th>Professional body</th>
<th>Patient/ consumer group</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>1.1 Perception of HGC consultation</strong></td>
<td>Did not know enough about consultation to fully comment but believed it had been fair. [AC1: S6: P47-49; AC2: S6: P62-64; AC3: S5: P89-91]</td>
<td>Only one interviewee of this group commented: that it was biased due to lack of public representation. [POL1: S7: P79, P87]</td>
<td>Lack of public representation but wide range of other stakeholders. Some felt that some members were biased against industry and decisions had been influenced by lobbying. [IND1: S5: P59, P63; IND2: S7: P40; IND3: S6: P48, P52, P69, P73; IND4: S5: P55, P76, P84]</td>
<td>Possible lack of representation from the public. Response to consultation based on views from individuals in organisation and small committees rather than more representative views. [PB1: S5: P65; PB2: S9: P94-96; PB3: S5: P26]</td>
<td>Believed consultation had been wide and fair. Responses to consultation not based on individual opinion. [PCG1: S6: p43, P52; PCG3: S2: P10-22]</td>
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<td></td>
<td>One participant felt that the consultation was based on suppositions and perceptions rather than real data. [PCG3: S10: P147]</td>
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<td></td>
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<td></td>
<td>One participant disagreed and felt that a lighter touch would have been more appropriate [PCG3: S8: P117]</td>
</tr>
<tr>
<td></td>
<td>Academic</td>
<td>Political/ regulatory body</td>
<td>Industry</td>
<td>Professional body</td>
<td>Patient/ consumer group</td>
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<tr>
<td><strong>2.0 Perception of industry sector</strong></td>
<td>General sense from most groups of mistrust towards the industry in terms of what they do with DNA samples, the accuracy of information they provide and their interest in selling genetic tests. [AC3: S3: P40; POL1: S3: P15, S5: P57, S8: P132; IND1: S2: P10; IND2: S12: P127; PCG1: S10: P95; PCG2: S2: P10, S3: P15, S7: P41]</td>
<td></td>
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<td></td>
</tr>
<tr>
<td><strong>3.0 Genetic test providers</strong></td>
<td>Uncertainty about pharmacists providing tests and advice; GPs better placed. [AC2: S7: P78; AC3: S6, P100, P104, P116] Belief that people will require advice from health professionals after receiving genetic information. [AC1: S3: P11; AC3: S6: P110-112]</td>
<td>One participant preferred tests to be available in doctors surgeries as opposed to supermarkets although pharmacists may be suitable. [POL1: S9: P139; POL1: S12: P184] Another participant felt that pharmacists would be well placed provided adequate training was involved. [POL3: S3: P16]</td>
<td>GPs do not have adequate level of understanding to provide tests [IND3: S10: P114; IND4: S8: P127]</td>
<td>Pharmacists not best placed to provide genetic tests and advice due to lack of time and knowledge, complex ethical issues to deal with, and GPs wanting responsibility for whole process. [PB1: S9: P150-P154; PB2: S9: P120-122]</td>
<td>One participant said that the vendor would depend on their knowledge and on the type of tests being sold. [PCG1: S10: P87]</td>
</tr>
<tr>
<td><strong>4.0 Consent</strong></td>
<td>General feeling that parents would only use a genetic test in the interests of their child. [IND3: S4: P26; IND4: S3: P34; PB1: S3: P40; PCG3: S5: P95]</td>
<td></td>
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<tr>
<td>5.0 Confidentiality</td>
<td>Academic</td>
<td>Political/ regulatory body</td>
<td>Industry</td>
<td>Professional body</td>
<td>Patient/ consumer group</td>
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<tr>
<td>Concern that genetic information would be used by police.</td>
<td>[POL1: S5: P53; POL3: S5: P37]</td>
<td>The individual owns their genetic information and should have a right to disclose it as he/she sees fit. Genetic data needs to be protected and kept confidential in order to avoid discrimination.</td>
<td>One participant felt that privacy of information was of paramount importance and that this was not possible if information was stored on NHS health records.</td>
<td>[IND1: S3: P23; IND3: S3: P12; IND3: S7: P61; IND4: S10; P140]</td>
<td>Concern about how private companies will handle genetic information. Concern about police having access to genetic data and how they will use it.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>6.0 Discrimination</th>
<th>Academic</th>
<th>Political/ regulatory body</th>
<th>Industry</th>
<th>Professional body</th>
<th>Patient/ consumer group</th>
</tr>
</thead>
<tbody>
<tr>
<td>Belief that insurance companies or employers will not discriminate on the grounds of genetic information as it will not be predictive enough.</td>
<td>[AC2: S4: P35, P72; AC3: S4: P65]</td>
<td>Belief that people will be discriminated against by the insurance industry on the basis of their genetic information.</td>
<td>Most genetic information is not useful at present to insurance companies, although public are concerned about it.</td>
<td>One participant felt that information concerning complex disease risk would be of no actuarial value.</td>
<td>Most genetic information will not be of use to insurance companies.</td>
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<tr>
<th>Academic</th>
<th>Political/ regulatory body</th>
<th>Industry</th>
<th>Professional body</th>
<th>Patient/ consumer group</th>
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</thead>
<tbody>
<tr>
<td>7.0 Genetic 'exceptionalism'</td>
<td>One participant felt that possible implications for family members on single gene disorder testing was the only distinguishing factor between genetic and other health information. [AC1: S4: P33] One participant highlighted that any medical test can be anxiety provoking, not just genetic tests. [AC3: S4: P63]</td>
<td>One participant did not think genetic information was any more predictive than family history. [POL1: S11: P149] One participant thought that genetic information can be perceived as more predictive but that in reality it is not. [POL2: S8: P96]</td>
<td>Belief that genetic tests are no different to any other medical test and should not be treated as any different. [IND1: S3: P19; IND2: S11: P84-86; IND3: S4: P25-26; IND4: S9: P135]</td>
<td>Genetic information is no more predictive than any other medical information, except in some cases such as highly penetrant disorders. [PCG1: S9: P81; PCG2: S7: P41; PCG2: S8: P46 PCG3: S5: P55] One participant expressed concern that imposing strict regulations would simply encourage the ‘genetic exceptionalism’ perception, already started by campaigning groups. [PCG3: S5: P72 PCG3: S10: P149-151]</td>
</tr>
</tbody>
</table>

8.0 Quality of genetic tests

General view that the quality – accuracy, reliability, validity – of the tests should be ensured. [POL2: S5: P50; IND2: S3: P12; PB3: S3: P11; PCG1: S2; P6]
<table>
<thead>
<tr>
<th><strong>9.0 Validity and usefulness of complex disease risk assessment</strong></th>
<th><strong>Academic</strong></th>
<th><strong>Political/ regulatory body</strong></th>
<th><strong>Industry</strong></th>
<th><strong>Professional body</strong></th>
<th><strong>Patient/ consumer group</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Questioned the usefulness of these tests in terms of predictive value – scientific evidence does not back them up.</td>
<td>These tests are only useful if treatment is available or if lifestyle changes are made.</td>
<td>Tests will have positive benefits as changing lifestyle could prevent illness.</td>
<td>Concerns around the evidence base but information should encourage positive lifestyle changes.</td>
<td>These tests have limited usefulness due to very low predictive value.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th><strong>10.1 Lay perception of genetics</strong></th>
<th>General view that there is a public lack of understanding about genetics and that the public perception is deterministic and negative.</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th><strong>10.2 Public understanding of information</strong></th>
<th>It is thought that the public generally misunderstand genetic information, particularly relative risk. More public education is needed.</th>
</tr>
</thead>
</table>
### 10.3 Impact of knowledge of genetics

<table>
<thead>
<tr>
<th>Academic</th>
<th>Political/ regulatory body</th>
<th>Industry</th>
<th>Professional body</th>
<th>Patient/ consumer group</th>
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<tr>
<td>[Referring to a complex disease susceptibility (CDS) test] Some participants felt that there was a danger of fatalism and believed people may become falsely reassured. However, another participant felt false reassurance was unlikely with low-moderate penetrance genes. [AC1: S3: P11; AC3: S3: P42] Some participants felt that genetic tests may be more anxiety provoking than other tests because of the perception of it being more predictive. [AC2: S3: P15; AC2: S4: P50; AC3: S4: P63]</td>
<td>[Referring to a CDS test and those for breast cancer] Concern about the emotional impact, causing depression or suicide. [POL1: S3: P15; POL2: S4: P33; POL 2: S8: P108; POL3: S4: P21]</td>
<td>One participant felt that the impact on other family members was important, particularly the issue of whether they should be informed of test results (if the test was for a condition that would have an impact on family), although another felt that the impact would depend on the individual and their life experiences. [IND2: S3: P12; IND2: S12: P155]</td>
<td>[Referring to a CDS test] One participant believed that there was no danger of fatalism if risks were adequately explained. [PB1: S3: P43]</td>
<td>Mixed views as to whether false reassurance, fatalism and excessive anxiety were valid concerns. [PCG1: S4: P26; PCG3: S5: P98]</td>
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[Referring to a CDS test and those for breast cancer] Concern about the emotional impact, causing depression or suicide. [POL1: S3: P15; POL2: S4: P33; POL 2: S8: P108; POL3: S4: P21]

Some participants felt that the impact on other family members was important, particularly the issue of whether they should be informed of test results (if the test was for a condition that would have an impact on family), although another felt that the impact would depend on the individual and their life experiences.

Mixed views as to whether false reassurance, fatalism and excessive anxiety were valid concerns.

One participant believed that there was no danger of fatalism if risks were adequately explained.

Mixed views as to whether false reassurance, fatalism and excessive anxiety were valid concerns.

One participant thought the information may encourage healthy behaviours.

Others mentioned the impact on other family members [referring to tests for inherited disorders] and the possibility that future gene-disease associations may have an impact if one already had knowledge of one’s genes. There was also mention of information encouraging illness prevention [referring to a CDS test]

[Referring to a CDS test] One participant thought the information may encourage healthy behaviours.

Others mentioned the impact on other family members [referring to breast cancer gene tests] and the possibility that future gene-disease associations may have an impact if one already had knowledge of one’s genes.

Others mentioned the impact on other family members [referring to breast cancer gene tests] and the possibility that future gene-disease associations may have an impact if one already had knowledge of one’s genes.

[PCG2: S3: P15; PCG2: S2: P10]
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<tr>
<th>10.4 Information, support and counselling</th>
<th>Academic</th>
<th>Political/ regulatory body</th>
<th>Industry</th>
<th>Professional body</th>
<th>Patient/ consumer group</th>
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<tr>
<td>One participant felt that tests for serious diseases should be accessed only where counselling is available.</td>
<td></td>
<td>Counselling or the information provided with the genetic test should explain results thoroughly.</td>
<td>Information should be meaningful and should explain what can be done to reduce risks of illness.</td>
<td>One participant thought that tests should be explained by an expert in case consumers have any queries; this would be unlikely via the NHS.</td>
<td>Concern that tests which are not offered through the NHS route will not provide adequate counselling, and explanations and time to reflect about the implications of the test.</td>
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<tr>
<th>11.0 Need for public education</th>
<th>Need for public education campaign to dispel misconceptions about genetics and allow test to be used appropriately.</th>
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| 12.1 Paternalism | It is wrong to prevent the public from having access to their health information and assume that they are not responsible enough to use a genetic test. | One participant felt that one reason for keeping access to genetic information in the NHS was that the medical profession did not trust patients to make good decisions on the basis of that information without them. | One participant believed that by imposing restrictions, the HGC are saying the public do not have a right to access their information. |
|------------------|-------------------------------------------------------------------------------------------------|----------|------------------|------------------------|
| [IND1: S2: P10; IND4: S6: P96] | [PB3: S8: P61-65, P69]                                                                         |          | [PCG3: S8: P117] |

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<tr>
<th>12.2 Personal freedom</th>
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<th>Industry</th>
<th></th>
<th>Professional body</th>
<th>Patient/ consumer group</th>
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<tr>
<td>Academic</td>
<td>Political/ regulatory body</td>
<td>Everyone should be free to access their personal genetic information, disclose that information to whomever they choose, have the freedom to choose where to access the information, and the freedom to choose whether they need counselling. [IND1: S3: P12; IND1: S11: P86; IND2: S11: P86; IND3: S3: P12, P16; IND3: S10: P109-114; IND4: S3: P28; IND4: S6: P96; IND4: S7: P102; IND4: S8: P114-118]</td>
<td></td>
<td>Need to strike a balance between personal freedom and protection from exploitation. [PB2: S10: P128; PCG1: S8: P71]</td>
<td></td>
</tr>
<tr>
<td>12.3 Protection of public</td>
<td></td>
<td>One participant believed that some members of the public would not understand genetic information and that there was potential for exploitation. [POL3: S10: P88]</td>
<td>Industry</td>
<td>Individuals should be protected from exploitation. [IND3: S8: P87; IND4: S10: P140]</td>
<td>One participant was in favour of protecting the public. [PB2: S3: P26]</td>
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<td></td>
<td></td>
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<td></td>
<td>One participant felt that there was not enough protection at present to allow the public access to genetic tests. [PCG1: S8: P71]</td>
<td></td>
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<tr>
<td>12.4 Responsibility for health</td>
<td><strong>Academic</strong></td>
<td><strong>Political/regulatory body</strong></td>
<td><strong>Industry</strong></td>
<td><strong>Professional body</strong></td>
<td><strong>Patient/consumer group</strong></td>
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<td></td>
<td></td>
<td>Would like to see more of an emphasis on preventative health care.</td>
<td></td>
<td>Adults are responsible for their own health.</td>
<td>[PB1: S7: P114; PB2: S10: P128]</td>
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<td></td>
<td></td>
<td>[IND1: S7: P82; IND2: S12: P119, P123; IND4: S2: P10]</td>
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<tr>
<td>13.1 Future</td>
<td>Some felt that there would be an increase in demand for genetic tests.</td>
<td>[POL2: S5: P42; PB1: S5: P69; PCG1: S10: P95]</td>
<td>Responsibility for health would increase.</td>
<td>[IND1: S7: P86; PB3: S10: P95-97]</td>
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<td></td>
<td>Tests would become more accurate in terms of prediction.</td>
<td>[IND3: S3: P12; PB3: S10: P85]</td>
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<td></td>
<td>There was a concern that it would be some time before the regulatory framework is put in place.</td>
<td>[IND4: S6: P64; PCG1: S7: P61]</td>
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<td></td>
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<tr>
<td>13.2 Uncertainty about future</td>
<td>Uncertainty about whether and how the tests will develop and how people will react to genetic information.</td>
<td>[IND2: S8: P69; PB1: S4: P48; PCG1: S7: P61; PCG3: S8: P117]</td>
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5.3.2 To what extent did stakeholders believe the public consultation to be 'comprehensive and objective'? Do stakeholders agree with the recommendations made by the HGC?

HGC consultation

Industry (IND) and professional body (PB) stakeholders and one political/regulatory (POL) participant felt that the public had not been represented well.

"I'm not sure the public at large had their say." PB1

In addition, it may be true that some organisations' consultation responses were not representative of their members. Some PB and patient/consumer group (PCG) participants reported that they had not consulted individual members or patients/consumers but had based their responses on the views of organisation committees or boards.

"The ... committee has been involved in these consultations so they've had an opportunity to bring their views in and they would hopefully represent a number of views ... from outside" PB2

5.3.3 What issues do stakeholders believe to be the most important and relevant to complex disease risk genetic testing and its access-related regulation?

Perception of industry sector

A general sense of mistrust towards ‘the industry’ was evident from the interviews. There was scepticism about the accuracy of information that the industry provides because of their commercial interest in the genetic tests.

"there’s too much hype. And let’s face it, it’s come from [the industry] who are trying to make money." AC3

"we’re talking about wealthy industries who are going to market these test products to sell them....I think that also links into creating demand for the tests." PCG1

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In addition, stakeholders seemed suspicious of how industries would handle consumer’s genetic information.

“drug companies might bribe somebody to give them that database once there’s 300,000 people on it.” GOV1

“[something] we don’t know about these companies – what are they doing with the information they get?” PCG2

This concern is also evident in members of the general public. Results from quantitative and qualitative research conducted in conjunction with the HGC consultation by YouGov (2003) and People, Science & Policy (2002) respectively indicated that the public would rather access genetic tests, information or results from a GP than by any other means. They would not trust commercial companies to sell a genetic test without there being a hidden motivation to sell another product on the back of it. In order to protect themselves, they believed private companies would be more likely to give out false positive rather than false negative results which would undermine the credibility and usefulness of the tests. There was also some suspicion about what companies would do with the DNA samples.

There is a body of literature attesting to the resistance of new technology. The general mistrust of industry by both stakeholders and the public could be related to this. New technology is resisted because risks are difficult to assess and this uncertainty prevents acceptance (Bauer 1993). Genetic technology is still relatively new and the potential problems and full impact of genetic information are still unknown. This can lead people to see it is as risky (Slovic 2001). Additionally, memories of the recent MMR vaccination controversy and GM crop debates may have tainted people’s perceptions of technology, science and industry, affecting their reaction to subsequent innovations.

Genetic test providers

There were mixed views on who would be best placed to provide genetic services. Academic (AC) and professional body participants tended to believe that GPs were
better placed than pharmacists because of their clinical knowledge and private consultation space.

"it shouldn't be the pharmacy; it should be the doctor because most of the drugs we’re talking about should be on prescription." AC3

This was also the view of some members of the public according to focus group research and a large internet survey on genetic testing (People, Science & Policy 2002; YouGov 2003). Accessing tests via the GP was thought to be preferable as they would have the patient’s medical history.

Industry participants however, felt that GPs did not have an adequate understanding of genetics to be able to provide comprehensive advice. Political/regulatory stakeholders tended to prefer health professionals in general as opposed to non-professional vendors such as supermarkets.

"the doctors I’ve met don’t know much about genetic tests and disease risks" IND3

Stakeholders did not tend to discuss any benefits of accessing tests through non-NHS routes yet People, Science & Policy (2002) reported that members of the public acknowledged that speed, convenience and confidentiality were advantages of buying tests privately. However, they also thought that restricting access to the NHS was important because of the potential impact of results on individuals, the impact of home testing on NHS resources and keeping genetic information on medical records.

The report following the HGC public consultation, Genes Direct was published in March 2003 (Department of Health 2003a). Based on the evidence and opinions it gathered during the consultation the HGC recommended that stricter controls be placed on the supply of genetic tests and testing services. It called for a “composite of controls based on general and specific legislation, regulatory bodies of several different complexions and a well supported series of Codes or guidance to ensure that the industry maintains high technical and ethical standards” (p.52).
The Commission drew a parallel with the way medicines are categorised into ‘prescription-only’, ‘pharmacy’ and ‘general sales’. They recommended that most genetic tests should not be available direct to the consumer, that is, bought from a retailer, via the internet or mail order, for use at home. Those tests which the public might in future be able to access without the need for health professional intervention would have to be demonstrably suitable and of high quality, both in terms of the test itself and the information and advice accompanying it. The provider would also need to be adequately trained.

The HGC based its conclusions on two main factors. First, the belief that consumers using genetic tests at home would be less likely to receive adequate information and support than consumers using tests via a health professional such as a GP or a pharmacist. Second, the danger that testing of non-consenting individuals would become more likely with an expansion of home testing.

At the time of writing the government had not yet officially responded to the HGC recommendations. They could take on the cautious advice from the HGC because in terms of public appearance, the government may believe that it is better to play safe to avoid disputes about risk exposure, even if it means delaying the opportunity for the public to potentially have information that may help prevent future ill health. In addition, evidence describing public views is scarce but those available suggest that the public are sceptical and concerned about new genetic technology and would rather it be over than under regulated (MORI 1999).

However, by protecting itself, this regulatory direction would also shelter citizens from new experiences, rather than exposure in a safe environment to allow familiarisation and progress. This is likely to prevent acceptance and understanding and reinforce the ‘scary’ and uncertain perception of genetics. In addition, it could discourage technological innovation in Britain, forcing the industry into other countries which have more relaxed regulations.

Alternatively, the government’s delay in responding to the HGC may be a tactic for allowing the technology to seep into the public consciousness, so that it becomes
more familiar and thus more acceptable, before deciding on a ‘lighter touch’ of regulatory policy.

Consent

There was a general feeling that consent in the case of children was a non-issue as parents would only use a genetic test in the best interests of their child.

"It's likely to be done in the child’s interest, it's another medical test.” IND3

One PB interviewee, however, tended to disagree with this view.

"A parent might think it might be quite interesting to find out if [their] child will develop Alzheimer's Disease...then the result of that could lie on that persons medical record and as they grew up they would have to deal with the consequences for that.” PB2

Another consent related issue was that of one’s DNA being tested for genes which had not been consented to. Although some participants emphasised that complex disease genetic tests would not be used to determine paternity others pointed out that testing for a gene linked to one disease may give information about the susceptibility to another disease.

"that's not what [complex disease risk] tests do...paternity tests are completely different.” PB1

"People consent to [a BRCA3 test] but what they don't consent to is if BRCA3...also increases your risk of schizophrenia quite substantially...You just need to think about what have people actually consented to.” PCG2

This was also linked with the availability of genetic tests over the counter.

"the problem with home testing is that it allows for things like taking a sample of somebody’s DNA without their knowledge or consent and using it perhaps for a different kind of test.” PB2
There are clearly different issues involved in the area of consent, some of which relate to different types of tests, some to the test provider and some to the access of testing services. This demonstrates the complexity of the area of genetic testing. Tests for differing purposes are likely to be used by different groups of individuals, will give different information, have different consequences and therefore may require different access restrictions. The need to consider varying types of test in their own context is therefore essential. However, in some stakeholders there was no clear sense of differentiation between tests.

Confidentiality

Many participants were concerned about the confidentiality of genetic information once a test had been taken. However, there were subtle differences between stakeholder groups. Both POL and PCG participants expressed concern about the potential for use by the police. This may be indicative of these groups’ fundamental wish to protect the public.

"what happens to that information anyway when you've got it - who gets hold of it?...It could end up in police hands."  POL1

Industry stakeholders were also concerned with confidentiality but believed that this could be ensured by keeping the data private and allowing the individual freedom to disclose it as he or she sees fit. This is an outward disclosure of a belief in individual rights but it may be masking the industry’s desire for tests to be available privately. However, one member of the professional body group also felt that privacy of genetic information was of utmost importance and believed that this could be ensured by not having one’s information stored on NHS records.

"as far as I’m concerned it’s just the individual and it’s then up to the individual who that individual gives access to...if you force someone to go through a gate keeper there are problems of confidentiality...I would rather have the right to go and do it without anybody knowing the results."  IND3

"I think, the most important thing to me personally is privacy, and this is the least private way [NHS], this is the least private."  PB1
A survey of public opinion revealed that some members of the public are also concerned about the confidentiality of genetic information (YouGov 2003). Of the survey participants who expressed scepticism about genetic tests for personal health, 17% were worried that the State may use their genetic information without knowledge or consent. The majority stated that they would not want companies or institutions such as employers, insurance companies, pharmaceutical companies, the government or the police to have access to their information. Results from focus group research confirms this (People, Science & Policy 2002). However, unlike the professional body representative above, the survey showed that only 10% of the public would not want the NHS to have access and only 4% would not want their GP to have access to their test information.

Discrimination

The use of genetic information to discriminate in insurance and employment situations was another scenario given prominence in the HGC consultation. However, four of the five stakeholder groups (AC, IND, PB and PCG) believed that information from most genetic tests (that is, information gained from complex disease risk tests) would not be predictive enough to be used usefully by insurers or employers.

"It actually won't happen. It's an issue, I understand. But I don't believe it will happen. Again because when people start thinking about it, they'll realise that genetic information isn't as predictive as it's thought to be." AC3

"We're a long way from the stage where the tests themselves would be able to give any information that could be so important to insurers and employers." IND3

"...most of the genetic information we're talking about here, much of it has very low predictive value and so if used rationally or actuarially, would have very few consequences." PCG3

This may have been a concern put forward by anti-genetics lobby groups, but which is, according to most stakeholders, a scenario which is actually unlikely to occur.
"I'm not sure the HGC themselves are that keen to make the link between the insurance debate and over the counter tests. I think it's just put to them all the time by campaigning groups." PCG3

POL participants, the only group to believe that genetic information will be used for discrimination, may be showing a protective nature towards the public.

"So you can quite easily, I think, see a situation arising where a person will simply be uninsurable." POL2

Findings from both the internet survey and the focus group research show that the public has strong views about not allowing employers or insurance companies access to genetic information (People, Science & Policy 2002; YouGov 2003). Unlike many of the stakeholders, it may be that the public are perhaps not aware that most genetic information will not provide any more information than that which insurance companies already ask for. Or, that if they are aware of this they maintain strong views about the ownership and privacy of that information.

Genetic 'exceptionalism'

Most participants across the board did not believe that genetic information was different to other medical information, in terms of either predictability or emotional impact. AC and PCG groups made the distinction between tests for single gene, highly penetrant disorders and tests for complex diseases.

"I don't see any reason why that should be different to any other test for any other biochemical marker or a pathological specimen." IND1

"it shouldn't be treated specially unless there are good reasons to" PCG3

One POL participant believed that the perception of genetic tests as opposed to other medical tests is different, but in reality it is not.
"I think it's scarier on an emotional level because it has this veneer of scientific validity.

POL2

This is consistent with the view that most genetic information will not be used for discrimination. However, some participants believed that the public still needs protection. The perception of public perception may be influencing opinion here. If stakeholders believe the public sees genetic information as special or different, perhaps they are more likely to give opinions on that basis, apparently representing public view. It is not clear exactly how the public feels about genetic information, however; stakeholders may be underestimating public understanding and perception. If this is true there is a danger that regulation will be formed around (perceived) public misunderstandings and perceptions rather than re-educating the public. Moreover, if participants believe that genetic tests are no more predictive than other medical information, should they be regulated in the same way as cholesterol or blood pressure tests, that is, available over-the-counter in a pharmacy? One PCG participant felt that imposing strict regulation might actually encourage genetic 'exceptionalism'.

Quality of genetic tests

The general view of participants was that it was important to ensure the quality of genetic tests. This encompassed the accuracy of the tests, how reliable the tests are in measuring what they purport to measure ('analytical validity') and the clinical utility of the tests (how useful and informative the test results are to the consumer),

"In terms of the main issues that would probably come to mind, I guess there's the issue of the efficacy of the tests and the usefulness of the tests themselves and their whole reliability so there's that whole quality of the tests." PCG1

Validity and usefulness of complex disease risk tests

This was a widely and strongly discussed issue across all stakeholder groups. Participants from the AC and PCG groups were concerned that there was not sufficient gene-disease association evidence to back up the claims of tests for
complex disease risk factors. Some of these participants held very negative attitudes towards these types of tests.

"I think that one of the dangers with the susceptibility tests is that the science is just not there just actually backing up what it is they really mean" AC1

"we share with many members of the HGC and many members of the professional genetic community quite a strong scepticism of the usefulness of some of the tests... I think a lot of the HGC and many professionals think these tests are a waste of time - they’re exploitative" PCG3

POL participants did not mention the evidence base, possibly because they are not aware of the issue. They tended to feel that the tests would only be useful if treatment for the diseases were available, or that lifestyle changes could be made to reduce risks. Both of these are true in the case of complex disease risk tests. This may indicate a positive attitude towards the potential of these tests.

"I would only have two concerns. One, was the test likely to be accurate, and two, could I do anything with the information? I don’t want to know anything about something I can’t do anything about. “ POL2

IND participants also did not mention the strength of the evidence base supporting these tests. It is unclear whether this indicates that they believe it is strong, or that they are not admitting that it may be weak. They strongly held that complex disease risk factor tests would have very positive benefits in terms of preventing future illness. PB participants also voiced concerns about the evidence base but were still positive about the tests’ potential benefit of changing lifestyle to prevent illness.

"the risk factor type cases, in which case people may be able to modify their lifestyles and so it, there may be some very positive benefits from undertaking such testing” IND2

"I don’t even know whether the data is there about the reliability of that information yet...I don’t think we’ve got the evidence” PB3

"I think people will change their behaviour” PB3
Regarding the evidence base for the tests, studies have yet to produce very consistent results about gene-disease associations due to differences in sample sizes and sample characteristics (Ioannidis et al 2003). However, large epidemiological research studies such as that by The Wellcome Trust Case Control Consortium (WTCCC) soon to be undertaken in the UK will provide more reliable information (Public Health Genetics Unit 2005).

A further point is that many other health products and treatments, such as homeopathy, are readily available to the public with weak supporting evidence for their health benefits. There may be evidence here of some participants treating these tests differently because they deal with DNA (also see genetic 'exceptionalism' below).

Public perception and understanding of genetic information

There was a general feeling across most stakeholder groups that the public perception of genetics is deterministic.

"people perceive genetics as being more predictive but its actually only slightly." AC3

"[the public think] OK, genetics, we're talking about something that's highly predictive"
PCG1

IND participants in particular raised the issue that the public also see genetic information as only giving bad news.

"I think people are very suspicious of it, they see it all as being bad news. Genetic testing can only either predict what you're going to die of and when, or it's going to give you bad news."
IND1

There was also a consistent view that the public does not understand the concepts of risk that are attributed to genetic information.
Chapter Five – Stakeholder analysis: Method, results and discussion

"people don't understand relative risk, they don't even understand percentages very much, a lot of people." GOV3

"the vast amount of the public have no concept of relative risk." PB3

These thoughts were linked to the view that more public education about genetics is required before this type of health information can be used in a positive and constructive way. Although stakeholders had little confidence in the public’s ability to comprehend risk information, they believed this could be improved with education.

"do people understand ... lower or higher risk? Maybe they don't in mathematical terms but we will have to find ways of putting that across in a common sensical [sic] kind of way and I think that can be done."
PCG3

"You can't just launch [genetic tests] onto the market - technology says yes you could because the products are there and they're available - but you actually need to educate the public first."
IND2

"what you've got to do is ... find ways of educating the public so that things are used appropriately."
PB3

Again, the stakeholders are showing a perception of public understanding of genetic risk information. This may have influenced their responses to the HGC consultation. On the other hand, focus group research suggested that stakeholders may be right. People, Science & Policy (2002) reported that participants generally thought genetics meant dangerous, serious and incurable. The perception of genetic diseases as incurable or unpreventable (even when facilitators suggested preventative action) affected the perceived value in getting tested for predispositions to illness. Participants were also unaware that spontaneous mutations, rather than inherited mutations, could occur and that DNA could be taken from sources other than blood. However, participants in another study seemed more confident in their knowledge of
It is argued that a basic understanding of science is necessary for society to interact with the many scientific and technological developments that impact on their lives (Sturgis, Cooper & Fife-Schaw 2005). Moreover, a knowledgeable citizen is essential in order for truly democratic decisions to be made. In a study by Durant, Evans & Thomas (1992) scientific knowledge was measured with a scale which taps both factual and theoretical knowledge and that of the rationale and processes underlying scientific enquiry. This scale has been found to be positively associated with self-reported interest in science as well as educational level, age, gender and social class. Using this scale, Durant, Evans & Thomas (1992) reported that the average score was less than 50% correct answers.

This may indicate a scientifically illiterate population and provides evidence for the 'deficit model' of public understanding of science. This model holds that resistance to science and technology is in part due to ignorance, superstition and fear. Better understanding of the science behind new technologies would lead to a higher acceptance of them (Sturgis et al 2005).

However, there is a debate around whether scientific knowledge can truly be measured. Some believe that public attitudes towards science are determined by 'institutional knowledge', the funding and regulation of science, whereas others believe understanding does not necessarily require knowledge (Durant, Hansen & Bauer 1999; Sturgis, Cooper & Fife-Schaw 2005). Others believe that the average citizen does not have the time or motivation to learn about scientific facts but that he uses everyday experience, values and interests to reach judgments (Sturgis, Cooper & Fife-Schaw 2005). Ascertaining attitudes towards science is also problematic given the diversity of subjects within that remit. Dietrich & Schibeci (2003) report an apparent hierarchy of acceptance, with technology that benefits organisms more likely to be accepted than that which simply improves the shelf-life of food or has cosmetic benefits. Therefore in order to fully understand attitudes towards science, specific areas must be examined.
There is evidence to suggest that a more knowledgeable public is more likely to find biotechnology applications morally acceptable and useful, and their risks tolerated (Gaskell, Allum & Stares 2003). However, other evidence suggests that although attitudes become more positive relative to increased knowledge this is related to the hierarchy of acceptance that beneficial applications may be appraised more positively, but reservations are increased for non-essential applications (Stratford, White & Park 2002; Sturgis, Cooper & Fife-Schaw 2003).

Impact of knowledge of genetics

This was an important issue for most participants in all stakeholder groups. However, details and views differed both between and within groups. POL participants expressed concern over the emotional impact of genetic information such as depression and suicide. There were mixed views as to whether miscomprehension of risk information could lead to false reassurance, fatalism or anxiety.

"the big issue with genetic testing...is the issue of false reassurance." AC3

"If somebody has a mutation they may see it as being much more fatalistic as opposed to having a poor lifestyle" PCG1

"these kind of predisposition genes...very rarely will it imply...a kind of fatalistic or a totally reassuring response." PCG3

Some mentioned the consequences of genetic information on other family members and others felt that there was a risk that future research would uncover more sinister links with the genes being tested at present; a knowledge that those who had undergone testing would have no control over.

"should other family members be informed? And they may not want to be informed, but they may...just because of the genetics of the particular disease, they may actually know without wanting to know their own information about themselves." IND2
“because science is developing so rapidly, it might be the case that that particular test has revealed other things about non-treatable disorders.” PB2

A few believed that genetic information might promote more healthy behaviours or illness prevention.

“if somebody realised that they might have a genetic susceptibility it may prompt them to take better care of themselves.” PCG1

In a focus group study some participants, particularly older men, did not think that they themselves would change their lifestyles (although they may take medicines), but acknowledged that healthier lifestyle changes would be made by some individuals (People, Science & Policy 2002). They also thought it was unlikely that people would want a predisposition test unless there was definite preventative action that could be taken as a result. This study reported no evidence that people thought false reassurance was likely.

Participants did tend to understand that the impact of information differs according to the type of genetic tests but occasionally they did not specify which type of test they were referring to when discussing this area. In terms of complex diseases, there are likely to be large differences in impact depending on which complex disease or illness the test is for. There are differing perceptions of seriousness, ability to prevent, treat or cure, and stigmas attached to illnesses such as cancers, cardiovascular disease and mental illness.

Information, support and counselling

Participants across all stakeholder groups agreed that the availability of advice and information accompanying genetic testing was necessary. Both POL and IND participants were concerned that the information provided was meaningful, with IND participants emphasising the need to explain what to do to reduce illness risks. One PB participant believed that an expert should explain information so that patients or consumers would have the chance to ask questions, both at the consultation and at
any time afterwards. She felt that it was unlikely that the NHS could provide such a service.

"I would still want to know that people had the access to good quality information in a way that they could actually understand it... that it's actually made meaningful for people so that they know what they can do to modify lifestyles." IND2

There is evidence that the public agree with this. The People, Science & Policy (2002) focus group research reported that the public did not expect genetic test results to be straightforward and thought they would require medical input to interpret results and advise on subsequent action.

PCG participants were concerned that services not offered through the NHS route would fail to provide adequate counselling, explanations or time to reflect about the implications of the test, indicating a mistrust of the private sector.

"How useful is that information to people who are quite desperate and anxious and often for no good reason and often good impartial information will be 'don't worry and don't waste your money on a genetic test'. That's not the information that they [commercial companies] give because they want to sell a test." PCG2

Need for public education

Some participants thought a public education campaign would be required to dispel any misconceptions about genetics and to allow test information to be used appropriately. One participant related this to the uncontrolled availability of information and services on the Internet and suggested that the only way preventing misuse was to educate the public.

"You can't just launch these onto the market - technology says yes you could because the products are there and they're available - but you actually need to educate the public first." IND2
"I do believe that to a certain extent you can't close the stable door so with the presence of purchase on the internet then what you've got to do is accept that and then find ways of educating the public so that things are used appropriately."

PB3

Paternalism, personal freedom, public protection and responsibility for health

IND participants felt that it was wrong for the government to prevent the public from having access to their own health information, and to assume that they would be unable to take the test responsibly. One PCG participant agreed; by imposing restrictions, the HGC were effectively saying that the public did not have a right to access their own information. There is a belief here that the government, or at least the HGC, are acting paternalistically about genetic information.

"I actually think it's wrong to say people are too stupid to take responsibility for their own tests or their own results for their own tests."

IND1

One PB participant believed that easier access to health information would lead to greater patient autonomy. However, he also believed that doctors would not trust patients to make suitable treatment decisions on the basis of that information. A possible underlying reason for restricted access to genetic information is that it would otherwise be a threat to medical expertise.

"I think the medical profession says well, no, patients can't make those decisions... they underestimate the ability of patients."

PB3

All IND participants felt strongly that individuals should be free to access and control their own personal genetic information. It was also mentioned by other participants in relation to striking a balance between freedom and protection.

"...anyone that wants to have information we should give them information"

IND4

"The problem is how you allow for more choice and autonomy for that group of adults while providing protection for other groups."

PB2
PB participants tended to believe that individuals are responsible for their own health yet were in favour of protecting the public and did not express strong feelings about personal freedom to information. Perhaps they do not fully trust the public to understand or use health information responsibly and are favouring public protection to avoid repercussions for the professionals they represent or to keep medical control in the hands of the professionals.

"Adults are responsible for their own health – absolutely." PB1

"[The] remit of this body centres on public protection…there would be lots of things that would need caution applying to them in terms of expanding the availability of genetic testing." PB2

Results from the focus group research revealed that the public were also concerned about the protection of vulnerable groups, specifically the elderly, those living alone and the ‘worried well’ who, it was thought, would be particularly susceptible to the marketing of genetic tests (People, Science & Policy 2002). However, when prompted by facilitators, focus group participants believed individuals had a right to their own genetic information.

**Future of genetics**

Most participants raised issues about various aspects of the future of genetics and genetic testing. There was some degree of uncertainty about how the tests will develop and how people will react to genetic information.

"nobody really knows how anybody’s going to really react to this sort of information." IND2

[referring to the HGC consultation] "I, really, our view is that it would have been best to just to see what happened a bit more - just give it a bit more time to see if these problems develop because in a sense what we’ve had is quite a heated debate about very little in a way so far about what might happen." PCG3
Chapter Five — Stakeholder analysis: Method, results and discussion

However, some felt that tests would become more accurate in terms of prediction of illness and disease and that there would be an increase in demand for genetic tests.

"I have no doubt that in time that will become refined so it is very very accurate...It's got enormous potential." PB3

"this is a big market already and it has all the signs of it being a bigger market." GOV2

These thoughts may be related to some participant’s views that responsibility for health and illness would increasingly be held by the individual.

"I think [the health care system is] moving to the preventative, there's no doubt about that....and I think [genetic tests for complex disease risk factors] will be one of a multiplicity of factors that allows them to do that." PB3

However, there was also concern among some participants that regulations may not be sufficient or able keep up with technological developments.

"we're more worried that this might be another issue that just falls through [a regulatory system] gap." PCG1

"it's going to be another several years before there is a solid regulatory framework that...the industry and the public can expect to see." IND4

During the interview, stakeholders were also asked to rate eight concerns or issues, which had been discussed during the HGC consultation, as low, moderate or high importance.

The Kruskal-Wallis non-parametric ranking test for independent groups was performed to identify how each of the stakeholder groups ranked the issues. Although there were no significant differences between stakeholder groups in how important the concerns were ranked, it appears that the academic group tended to rank all issues as lower importance than the other stakeholder groups, as indicated by a lower mean rank (see Table 5.4). As arguably the most knowledgeable in terms of
the biology of genetics this group may be being realistic about which concerns are more or less likely to be realised. In contrast, patient/consumer groups tended to rank issues higher, perhaps due to their role of protecting the public from any possible adverse consequences.

Table 5.4: Mean rank and sum of ranks of issues rated as important

<table>
<thead>
<tr>
<th>Issue</th>
<th>Academic</th>
<th>Political/ regulatory body</th>
<th>Industry</th>
<th>Professional body</th>
<th>Patient/ consumer group</th>
<th>Sum of ranks (all stakeholder groups)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Test information could be used to determine or trace paternity</td>
<td>3.83</td>
<td>7.83</td>
<td>9.50</td>
<td>8.00</td>
<td>8.00</td>
<td>33</td>
</tr>
<tr>
<td>Test information might not be accurate, and/or statements about error rates might not be understood</td>
<td>4.17</td>
<td>9.50</td>
<td>9.50</td>
<td>7.33</td>
<td>9.50</td>
<td>40</td>
</tr>
<tr>
<td>Test information could be used to determine or trace individual identity</td>
<td>6.17</td>
<td>9.50</td>
<td>7.00</td>
<td>6.50</td>
<td>8.50</td>
<td>32</td>
</tr>
<tr>
<td>Test information might cause anxiety</td>
<td>3.00</td>
<td>8.67</td>
<td>7.50</td>
<td>11.00</td>
<td>11.00</td>
<td>38</td>
</tr>
<tr>
<td>Test results may lead to discrimination and/or a 'genetic underclass' if disclosed to insurance companies or employers</td>
<td>6.83</td>
<td>9.00</td>
<td>7.00</td>
<td>9.00</td>
<td>5.75</td>
<td>38</td>
</tr>
<tr>
<td>Test information might evoke false reassurance about health</td>
<td>5.17</td>
<td>9.00</td>
<td>5.63</td>
<td>10.00</td>
<td>10.00</td>
<td>29</td>
</tr>
<tr>
<td>Tests could be done on children or others unable to give informed consent</td>
<td>7.50</td>
<td>8.17</td>
<td>4.50</td>
<td>11.50</td>
<td>8.50</td>
<td>32</td>
</tr>
<tr>
<td>Test information might cause people to become fatalistic about their health</td>
<td>8.00</td>
<td>8.00</td>
<td>8.00</td>
<td>6.17</td>
<td>10.75</td>
<td>30</td>
</tr>
</tbody>
</table>
The sum of ranks in Table 5.4 shows which issues tended to be rated higher than others (indicated by a higher number). The issue rated most often as highly important was that test information might not be accurate or may not be understood. This widespread concern corresponds with the interview data where most participants believed that the public misunderstand genetic information, particularly the concept of relative risk, and that public education and the availability of advice and information accompanying genetic tests is essential.

This may be related to another highly ranked issue; the belief that test information might cause anxiety. This corresponds with interview data where some participants believed that the anxiety-related impact of test results was a valid concern.

That test results may lead to discrimination was another issue that was rated consistently as highly important. However, the interview data indicated that the majority of participants believed that most genetic test information would not be predictive enough to be of actuarial value. It may be that stakeholders believe the public are concerned about this possibility and therefore rate it as a highly important issue, despite having a personal belief that it will not happen in reality.

That test information could be used to trace individual identity or paternity tended to be ranked as neither high nor low importance. However, these were not issues which were raised as important during interviews. Similarly, concern that tests could be done on those who had not, or were unable to, give informed consent was not ranked overall as highly important. This is compatible with the interview data where there was a general opinion that parents would only use a genetic test in the interests of their child.

The possibilities that test information could cause false reassurance or fatalism were ranked the least important issues compared to others. This may reflect the findings of the interview data, which showed mixed views on this issue.
5.3.4 Do stakeholders' views on individual autonomy over health information, and individual responsibility for health, differ when acting as representatives of their organisation than those they hold as individuals?

Participants marked their opinions on visual analogue scales (see Figure 5.1). They were asked to mark their view firstly as a representative of their organisation or profession, and then as an individual. The scales were from 1 to 10, although this was not explicitly indicated. By measuring the distance of the mark from the left, a quantitative value of between 0 and 10 was ascertained. Median values for these scores were then calculated where a lower score indicates agreement with responsibility for health and free access, and a higher score represents views which lean towards public protection and professional direction (see Table 5.5).

Table 5.5: Median values of visual analogue scales

<table>
<thead>
<tr>
<th></th>
<th>n</th>
<th>Median value of all groups</th>
<th>Median value for each stakeholder group¹</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>AC</td>
</tr>
<tr>
<td>Responsibility for health vs. professional direction*</td>
<td>14</td>
<td>1.75</td>
<td>3.50</td>
</tr>
<tr>
<td>Free access vs. public protection</td>
<td>15</td>
<td>3.00</td>
<td>6.00</td>
</tr>
<tr>
<td>Responsibility for health vs. professional direction*</td>
<td>13</td>
<td>3.50</td>
<td>6.25</td>
</tr>
<tr>
<td>Free access vs. public protection</td>
<td>14</td>
<td>4.50</td>
<td>6.75</td>
</tr>
</tbody>
</table>

¹AC Academic; POL Political/regulatory body; IND Industry; PB Professional body; PCG Patient/consumer group.

* Responses to statement as an individual and as an organisation significantly different (z=2.194, p=0.02)

Table 5.5 shows that, for the whole sample, the median values for the scales marked as a representative of an organisation are higher than those marked as an individual.
Results of a Wilcoxon Signed Ranks test for repeated groups revealed that the opinions about 'responsibility for health versus professional direction' stated as representative of an organisation were significantly different to those stated as a personal view ($z = 2.194, p=0.02$). In addition, this effect approached significance in the 'free access versus public protection' scale ($z = 1.844, p=0.07$).

This indicates that participants may have differing personal views to those they express on behalf of their organisation. Specifically, that they tend to personally believe that individuals are responsible for their own health and that they should have free access to their own health information, yet when speaking on behalf of their organisation they hold a more protective view, that the public needs protection and professional direction.

A phenomenon found in social psychology research has shown that group decisions about risk taking can be more risky or more cautious than decisions made individually (Stoner 1968). One explanation for these 'choice shifts' is that, when in groups, individuals make decisions based on perceptions of the widely held opinion (Eliaz, Ray & Razin 2004). If the widely held opinion favours a cautious decision, individuals will tend to be cautious. They will consider themselves to be more cautious than people similar to themselves, and group discussion will lead them to prefer still more cautious decisions. It is thought that this group shift is due to the group discussion increasing the salience of the widely accepted views and/or confirming the desirability of being cautious (Stoner 1968).

In the context of the HGC consultation, stakeholders may have responded in the way that they felt others would respond; they may have been reluctant to agree with more 'risky' regulatory strategies if they thought they would be alone in this decision. If the group discussion confirmed generally held cautious views then the group decision would be for a stricter regulatory position.

Another explanation for this choice shift is that it might, for example, be regarded as being against a charity's or professional body's interest to publicly be seen to be taking a 'permissive' view. They may run the risk of being charged with advocating action which might lead to easily identifiable harm even in just a very few cases.
Harm caused by unduly precautionary approaches is normally much more difficult to render visible.

Or, it could be because participants believed that they were more knowledgeable or understanding about genetic testing and its implications due to the nature of their job than the members or individuals that the organisations represent. This relates to the evidence found in the interview data that participants tended to believe that the public do not understand genetics or genetic risk information. An alternative reason for the discrepancy could be that the organisations are being over-cautious about the negative aspects of public access to genetic testing. This implies over-protection by the experts - a ‘better to be safe than sorry’ attitude towards regulation - which would hinder public access to genetic services. However, this phenomenon is even true of industry participants who were much more in favour of free access and individual responsibility.

The only stakeholder group not to exhibit this trend was the political/regulatory group. They may be reflecting the present political agenda towards increased individual responsibility in terms of health characterised by recent programmes and documents such as The Expert Patient (Department of Health 2001) and Choosing Health White Paper (Department of Health 2004b), although there is no evidence in the interview data to support or challenge this.

The choice shift finding prompted further investigation of the interview data. The visual analogue scale scores (individual views) were summed and low or high scorers were identified. Participants with low scores would be those who tended to agree with the free access of information and individual responsibility for health. Those who scored highly tended to agree towards the other end of the dimension, that the public needs professional control and protection. Interview data for low and high scorers was compared. However, no obvious differences in discourse or attitudes were seen.

If this trend for discrepancies between personal and organisation views were true across all the stakeholders involved in the HGC consultation there would be important implications. Although the individuals who responded to the consultation
may have a wide knowledge of the area, it is not necessarily true that members of the public are not capable of understanding this knowledge. At best this is over-protection and at worst, patronising. If participants were giving advice and recommendations based on perceived knowledge and understanding of the public, members or patients, the eventual recommendations for regulation would not be based on a representative opinion of the general public. Indeed, if this is a common pattern for the consultation process as a method of gathering opinion, it is seriously flawed. The interview data provides some support for this, as discussed above: some participants felt that the public had not been represented well in the HGC consultation and that some consultation responses were based on committee members' opinions, rather than a representative view of the professionals, patients or public they represented.

5.3.4.1 Views on wider issues and implications of access regulation

Participants were also asked to rate their agreement on a Likert scale from Strongly Agree to Strongly Disagree to twelve statements relating to various issues involved in the debate. Table 5.6 shows a frequency count of participants completing this item (please see Appendix, page 261 for full statements).
Table 5.6: Frequency count of participants' agreement or disagreement to statements, with median score of items highlighted.

<table>
<thead>
<tr>
<th>SUMMARY OF STATEMENT</th>
<th>Strongly agree</th>
<th>Agree</th>
<th>Neutral</th>
<th>Disagree</th>
<th>Strongly disagree</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Increase in NHS resource demands if free access</td>
<td>3</td>
<td>7</td>
<td>3</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>2. Counselling for complex disease tests is essential</td>
<td>10</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>3. Restrictive regulation may drive entrepreneurs out of the UK</td>
<td>3</td>
<td>3</td>
<td>2</td>
<td>8</td>
<td>8</td>
</tr>
<tr>
<td>4. Changing health behaviour is a good reason to allow free access</td>
<td>3</td>
<td>5</td>
<td>3</td>
<td>5</td>
<td>5</td>
</tr>
<tr>
<td>5. Public won't understand complex disease genetic test information</td>
<td>4</td>
<td>1</td>
<td>8</td>
<td>3</td>
<td>3</td>
</tr>
<tr>
<td>6. Advertising Prescription Only Medicine is undesirable</td>
<td>5</td>
<td>6</td>
<td>3</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>7. Internet access makes strict regulation impractical</td>
<td>1</td>
<td>1</td>
<td>12</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>8. Pharmacies are better placed to sell complex disease tests than GPs</td>
<td>5</td>
<td>5</td>
<td>5</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>9. Some populations are vulnerable to exploitation</td>
<td>3</td>
<td>3</td>
<td>5</td>
<td>5</td>
<td>5</td>
</tr>
<tr>
<td>10. Strong evidence base is vital (n = 15)</td>
<td>4</td>
<td>8</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>11. Too much emphasis on regulation</td>
<td>3</td>
<td>2</td>
<td>5</td>
<td>4</td>
<td>2</td>
</tr>
<tr>
<td>12. People have a right to access information</td>
<td>5</td>
<td>8</td>
<td>3</td>
<td>3</td>
<td>3</td>
</tr>
</tbody>
</table>

There are mixed responses on several statements. Fairly equal numbers of participants agreed, disagreed or were neutral with the statement that behaviour change would be a good reason to allow free access to genetic tests. This corresponds well with interview data where there were mixed views as to whether genetic information would prompt healthy behaviour change. In addition, there were equivocal responses to the statement of whether pharmacies would be best placed to sell complex disease genetic tests. Similarly, the interview data shows varied opinions on this issue with some participants favouring pharmacies and some favouring GPs. The issues of the exploitation of vulnerable populations and an overemphasis on regulation were issues which were not raised during interviews and provoked equivocal responses when posed as a statement.
Participants tended to agree that there would be an increase in NHS resource demands if genetic tests were freely accessible. This issue was not spontaneously raised during interviews, which suggests that it was perhaps not regarded as important as other issues. There was also general agreement that counselling for complex disease tests is essential. This view was apparent in interviews from participants across all stakeholder groups. The agreement that a strong evidence base for genetic tests is essential was confirmed by the interview data where many expressed concerns about the weak evidence behind tests for complex disease risks. Most participants agreed that people have a right to access health information. This view was also expressed during interviews although some participants mentioned it together with a need for public protection thereby giving an impression of preferring 'restricted' free access. The statement relating to the undesirable advertising of Prescription Only Medicines was included as a medicines-related information issue. Most participants agreed with this statement, giving the impression that the public are not capable of coping with freely available medicines-related information. This may also be an indication of views towards other health-related information.

Statements reporting that restrictive regulation may drive out UK entrepreneurs and that internet access makes strict regulation impractical tended to be disagreed with. These issues were not raised spontaneously during interviews. Whether this was because they were not thought to be important issues, or whether they had not been considered previously by participants is not known. These responses and relative lack of discussion may indicate an inability to see the wider impact of the regulation of access to genetic tests. Over regulation may discourage businesses and industries to settle and prosper in the UK. This would have both economic and social impacts, such as revenue and employment opportunities. In addition, the UK would never hold a 'cutting edge' position, as it would simply track developments and consequences in other, less regulated, countries until it was confident of outcomes. Although this may be a safer way to operate, potential health benefits may be lost. The government has pledged to invest £50 million in genetic technology, laboratories and workforce (Department of Health, 2003b) yet if they do not support the development of the industry in the UK, the profits of this investment will be enjoyed elsewhere.
Participants tended to disagree with the statement that the public would not understand complex disease risk information. This contradicts what they expressed during interviews. Most felt that the public see genetics as pessimistic and deterministic and have trouble understanding risk information. It is unclear why participants gave conflicting views although it may be due to the nature of the questioning. During interviews the concept of public understanding of genetics and risk information was discussed without prompting from the interviewer. In this sense it would be expected that these views were genuine. However, when asked directly whether they agreed with a statement, participants were less reluctant to express these views. It may be that stakeholders personally believe that the public are not capable of understanding this type of information but they are less willing to express this view to the interviewer.

Spearman’s rho non-parametric correlations were performed on the responses to these statements to investigate possible associations between views and attitudes. Although the sample size was small, the correlations gave an idea as to which views may be linked and prompted further investigation. Responses for ‘strongly agree’ through to ‘strongly disagree’ were recoded from 1 through to 5 on SPSS. Table 5.7 shows the strong correlations which emerged from the data.
Stakeholders who tended to agree that the possibility of changing health behaviour was a good reason to allow free access tended to disagree that free access to genetic tests may increase NHS resource demands (rho = -0.70, p=0.004). It is possible that these stakeholders are in favour of individuals taking more responsibility for their health and see genetic tests being used to motivate lifestyle changes without the need for the NHS.

Those who tended to disagree that Internet access makes strict regulation impractical were more likely to disagree that the public would not understand complex disease genetic test information (rho = 0.66, p=0.008). It is likely that this correlation is strong because most stakeholders disagreed with these two statements. It is interesting however, that in this style of questioning the majority felt that the public would understand complex disease risk information, yet the interview data suggests the opposite.
Stakeholders who agreed that there was too much emphasis on regulation were more likely to agree that people have a right to access information (rho = 0.76, p=0.001) and that restrictive regulation would drive out entrepreneurs (rho = 0.56, p=0.029). They also tended to believe that NHS resource demands would not increase if free access is allowed (rho = -0.62, p=0.013), that counselling for complex disease risk tests is not essential (rho = 0.58, p=0.023), and that some populations are not vulnerable to exploitation (rho = 0.73, p=0.002).

It could be argued that these views all fit with an industry position. Those who tended to agree that people have a right to access their genetic information also agreed that restrictive regulation might drive entrepreneurs out of the country (rho = 0.79, p=0.00). Again, this is likely to be an industry viewpoint.

However, there may also be some who believe that there will be an increase in NHS resource demands should free access be allowed, that counselling for complex disease risk tests is essential, and that there are some individuals who would be vulnerable to exploitation. They would also disagree that restrictive regulation would drive new business out of the UK. It could be argued here that patient/consumer groups may hold this position.

Stakeholders who agreed that Prescription Only Medicines should not be advertised tended to disagree that regulation might affect industry in the UK (rho = -0.62, p=0.013). This result may be a function of the majority of participants expressing these views.

5.4 Conclusions

- Stakeholders generally believed that genetic tests for complex disease risk factors were no more predictable than other medical information and would not be predictive enough to be used for discrimination purposes, although still ranked it as an important issue. It is possible that they are responding in the way they perceive the public view this issue.
Chapter Five — Stakeholder analysis: Method, results and discussion

- Stakeholders generally believed that the public perceived genetics to be deterministic, giving only bad news and that they were not capable of understanding genetic or risk information. Other evidence suggests that although the average citizen may not have factual scientific knowledge, everyday experiences are sufficient to reach judgements (Sturgis, Cooper & Fife-Schaw 2005).

- There were mixed views as to whether genetic test information would lead to anxiety, fatalism or false reassurance. Other evidence suggests that there are diverse opinions amongst the public too (People, Science & Policy 2002).

- Stakeholders tend to personally believe that individuals are responsible for their health and should have free access to their own health information, yet when speaking on behalf of their organisation they hold a more protective view, that the public needs protection and professional control. This 'choice shift' is a well documented phenomenon (Eliaz et al 2004) and could be due to their belief that the public do not understand well genetic or risk information and are acting paternalistically. This finding has implications both for the regulation of genetic testing services and for the validity of public consultation exercises.

The next chapter will present and discuss the results of the experimental questionnaire study, which aimed to assess the impact of genetic tests for complex disease risk factors.

5.5 Limitations of the study

Qualitative analysis allows an in-depth exploration of a subject area. Small sample sizes are often used because of the level of detail in the analysis. Whilst one may find patterns of attitudes across the sample one should not presume that these findings are true in other samples, situations or points in time. These findings provide a snapshot of opinions within this sample, related to the HGC consultation on public access to genetic testing in June and July 2003.
Although the sample size was appropriate for qualitative analysis, it was not ideal for the quantitative measures. While these scales were useful to give an indication of trends one cannot generalise these findings more widely.

The issue of subjectivity in qualitative analysis is ubiquitous. The researcher inevitably has preconceived ideas and although a proportion of the analysis was checked and confirmed by a second researcher, and annotated transcripts provide a transparency to allow scrutiny, there may still have been some bias in the interpretation of the data.

The method used to collect data in this study was a semi-structured interview, primarily because there were specific views to be elicited. Although free discourse was encouraged to some extent a less structured method of interviewing may have elicited views and experiences which were not revealed in the present study.

In terms of equality of participants, although all those interviewed had knowledge of the issues surrounding the HGC consultation on access to genetic testing services, not all had been officially involved. It is probable that some participants therefore had more knowledge or insight into this subject than others. Additionally, some interviews lasted more than twice as long as others due to participants' time constraints. This inevitably led to more detailed discussion from the longer interviews. Issues which were not referred to in shorter interviews may have been eventually been discussed if the interview had lasted longer.
CHAPTER SIX – EXPERIMENTAL QUESTIONNAIRE STUDY:

Methods, Results & Discussion
6.1 Introduction

The aim of this study was to investigate the potential value of genetic tests for complex disease risk factors in addition to individuals' concerns about the use of genetic information. This chapter explains the methods used to accomplish the experimental questionnaire study and fully describes the analysis and results. It discusses the findings in the context of other research and considers its implications. Limitations of the study are presented and future areas for investigation are proposed.

6.2 Method

6.2.1 Designing the questionnaire

The questionnaire for this study consisted of previously validated instruments and items designed for the purposes of this study. The validity and reliability of the instruments and items are detailed below.

*Multidimensional Health Locus of Control Scale (MHLC: Wallston, Wallston & DeVellis 1978)*

Individuals are proposed to have an internal or external locus of control where some attribute personal responsibility for events or behaviours and some attribute responsibility to luck, fate or important other individuals. Health locus of control is a construct specifically related to health behaviours (for more detail, see Section 2.1.2). The MHLC scale contains six items each relating to internal, external and powerful others locus of control. Original internal reliability was good ($\alpha=0.77$, $\alpha=0.67$, $\alpha=0.75$ for internal, external and powerful others scales respectively) and subsequent studies have confirmed this (e.g. $\alpha=0.86$, $\alpha=0.77$, $\alpha=0.62$ respectively: Schroder & Schwarzer 2005).

The MHLC was criticised by Rogers (1995) for not allowing for a concept of self-blame and divine influence. Wallston agreed and the God Locus of Health Control (GLHC) was subsequently developed to be used as an adjunct to the MHLC (Wallston et al 1999). However, the GLHC has been little used; the majority of
studies use only the MHLC. Considering this, and the length of the questionnaire, it was decided not to include this extra scale.

The MHLC scale was validated on members of the public (Wallston, Wallston & DeVellis 1978) and has been subsequently validated on outpatients (Marshall, Collins & Crooks 1990). The latter authors also compared the MHLC with another health locus of control measure, the Lau-Ware HLC scale (Lau 1982). Cronbach’s alpha coefficients for the Lau-Ware subscales were unacceptably low and factor analysis failed to replicate its dimensions. The psychometric superiority of the MHLC was the reason for choosing to use this instrument. Form A of the MHLC was chosen because it is equivalent to Form B but is generally used in a healthy sample (Wallston 1993). Form C of the MHLC is used in samples with medical conditions as it can be individualised to a particular illness.

Health Value Scale (Lau, Hartman & Ware 1986)
Wallston (1992) argues that internal locus of control will have a stronger influence on health behaviour among individuals who value their health highly compared with those who value it below other priorities (for more detail, see Section 2.1.2). The Health Value Scale reported an internal reliability of $\alpha=0.67$ in undergraduate students.

There are other scales which have been developed to measure health value. The Rokeach Value Survey (Rokeach 1973) asks participants to rank in order of importance a series of values including health. This scale was not chosen for this study as it has more items than Lau et al’s (1986) and is cited less frequently in the literature. Similarly, the Health Value Index by Seeman & Seeman (1983) has not been used as extensively as Lau et al’s (1986).

6-item Short-Form State Scale of the State-Trait Anxiety Inventory (Marteau & Bekker 1992)
The State-Trait Anxiety Inventory (STAI) is a well established measure of anxiety. This short-form of the state scale has six items as opposed to 20 items in the full version yet it correlates $r=0.95$ with the full version. It includes three anxiety-present
and three anxiety-absent items which have shown internal reliability of $\alpha=0.82$ (Marteau & Bekker 1992). This scale has been used successfully in diverse areas such as desire of acutely unwell patients to participate in discussion about resuscitation (Fidler, Thompson, Freeman et al 2006), diabetes screening anxiety (Skinner, Davies, Farooqi et al 2005), and subjective stress in spinal cord injury patients (Rintala, Robinson-Whelen & Matamoros 2005). The scale was included in the questionnaire due to its conciseness.

**Nutrition-specific Self Efficacy Scale (Schwarzer & Renner 2000)**

Self-efficacy is the confidence one has of carrying out a behaviour (for more detail see Section 2.1.2). This scale was designed to measure self-efficacy specifically in relation to nutrition and diet and was designed with other health-specific self efficacy scales. It consists of five items for which internal reliability has been reported as $\alpha=0.87$ (Schwarzer & Renner 2000). This is the only self-efficacy scale found in the literature to be specifically targeted at diet.

**Miller Behavioral Style Scale (MBSS: Miller, 1987)**

The MBSS measures an individuals’ dispositional coping style (for more detail see Section 2.1.3). Four scenarios are each followed by eight statements detailing possible response behaviours. Participants are asked to mark which behaviours they would be likely to perform in response to each given scenario. Half of the 32 behaviours are ‘monitor’ style responses, and the other half are ‘blunter’ style responses.

Some studies have reported the internal reliability of the MBSS (Miller 1987). Cronbach’s alpha coefficients for ‘monitor’ items has been reported as $\alpha=0.79$ and $\alpha=0.75$, and for ‘blunter’ items as $\alpha=0.69$ and $\alpha=0.67$. The scale has also been independently validated by factor analysis on 583 undergraduate students (Muris & Schouten 1994). Populations on which the MBSS has been successfully used include students, cancer patients and African-American women in a breast self-examination programme (Jacob, Penn, Kulik & Speith 1992; Steptoe 1989).
Risk perception

Risk perception was scored on a visual analogue scale measuring 10cm long on the questionnaire with Very Low at one end, Very High at the other, and Average indicated in the middle. Participants were asked to mark on the scale in answer to the question "What do you think your risk of developing heart disease is, compared to the average person the same age and gender as you?". The mark was measured with a ruler so that scores could range from 0 to 100 (millimetres). Previous studies have used similar single-item scales. Yanushka-Bunn et al (2002) measured perceived susceptibility with a single item which asked respondents to indicate on a scale of 0 to 10 the extent to which they thought they were likely to get colon cancer in their lifetime. Bosompra et al (2000) also used a single item to measure the extent to which respondents thought they were likely to get cancer in their lifetime.

Intention to change diet

As with risk perception, intention to change diet was scored on a visual analogue scale measuring 10cm long on the questionnaire with Very Unlikely at one end and Very Likely at the other. Participants were asked to mark on the scale in answer to the question "How likely is it that you will change your diet to help lower the risk of heart disease?". It was measured with a ruler so that scores could range from 0 to 100 (millimetres).

This ‘self-prediction’ measure has been shown to be more strongly related to actual behaviour than a more direct “I intend to perform this behaviour”-type measure (Sheppard, Hartwick & Warshaw 1988). The former is thought to include consideration of facilitating or inhibiting factors to the performance of the behaviour (Armitage & Connor 2001)

Health beliefs

As an important component of behaviour (see Section 2.1.2) three items measuring health beliefs were included in the questionnaire. The items were not based on any previous scale or measure and were intended to give a general indication of individuals’ health attributions rather than an in-depth insight into health beliefs.
Self-rated health
Self-rated health has been shown to be an independent predictor of mortality, possibly because it influences health-related behaviours which subsequently affect health (Idler & Benyamini 1997; Moller, Kristensen & Hollnagel 1996) (see Section 2.1.2).

Self-rated health was measured using a single item with a response scale of five points: "How would you rate your overall health?". A review on the association between self-rated health and mortality reported that most measures of self-rated health were designed as such (Idler & Benyamini 1997).

Fruit/veg & sugar/fat intake
Intake of fruit and vegetables was measured using a four-point scale of 'less than 2', '2 or 3', '4' or 'at least 5'. Sugar and fat intake was measured using a five-point scale from 'hardly ever' to 'every day'. These measures were taken as a general indication of diet and were not based on any previous items.

Activity level
Level of physical activity was scored on a visual analogue scale measuring 10cm long on the questionnaire with Not at all active at one end and Very active at the other. Participants were asked to mark on the scale in answer to the question "How active would you say you are?". It was measured with a ruler so that scores could range from 0 to 100 (millimetres). This was not based on any previous scale.

Smoking status
Participants were asked to answer Yes or No to the question "Do you smoke?"

The MTHFR gene
The MTHFR gene was chosen as the basis for the hypothetical test for two reasons. First, it is a real gene which has been linked relatively consistently with a complex condition (heart disease) (eg. Andreassi et al 2003) despite evidence that many findings from gene-disease association studies are not replicated (Ioannidis et al 2001). Second, the heightened risk of heart disease due to the interaction between the MTHFR gene and homocysteine levels can be reduced by altering the diet (Liu,
Chiang & Chen 2004). The aim of the study was to measure the impact of genetic test information (the MTHFR gene) on behaviour (diet) and so the MTHFR gene was a fitting example.

**Nutritional information**

The recommended daily intake (RDI) of folic acid was set at 600 micrograms (µg) for participants who had the MTHFR variation. This was a mean average of the RDI for MTHFR homozygotes at 800µg, and the RDI for MTHFR heterozygotes at 400µg (personal communication, 2004), since the hypothetical test results did not specify whether participants were heterozygous or homozygous for the mutation. Participants who did not have the mutation were recommended 400µg per day. Although the UK and EU recommendations are 200µg for an adult (Mason 2001) it is widely accepted by nutritionists, and indeed, is the RDA in the US, that 400µg is more suitable (personal communication, 2004).

**Experimental conditions**

In order to assess the impact of genetic risk information two types of test result were designed. One gave a 'positive' result which told of an increased risk of heart disease due to a MTHFR variation. The other gave a 'negative' result which informed of a lack of the MTHFR variation and therefore no increased risk for heart disease. Within the 'positive' results there were two further sub-conditions. These manipulated the framing of the message (See Section 2.1.4). The information reporting that changing the diet to include more folic acid would affect the risk of heart disease was either positively framed or negatively framed. The positively framed information read "If you change your diet to include more of these foods you are likely to reduce your risk of heart disease." The negatively framed information read "If you do not change your diet to include more of these foods you are likely to have an increased risk of heart disease." Therefore there were three test results that participants could receive:

---

1 information supplied by a nutrition consultant for Sciona 18/02/04
Concerns about, and comprehension of, genetic information

Based on issues raised during the HGC consultation concerns about the use of genetic information by the government, insurance companies and employers was measured using a 5-point Likert scale. In addition, the extent of comprehension of the genetic information given in the questionnaire was also measured using a 5-point Likert scale. These items were not based on any previous measures.

6.2.2 Piloting the questionnaire

Firstly, the genetic and test results information was piloted on nine individuals of varying ages, education and gender. The information was altered slightly following comments but overall the information was understood well by all.

The complete questionnaire was then piloted on administrative and teaching staff at a London university. The essential purpose of this pilot was to test the logistics of administering the questionnaire electronically and by post. Distributing questionnaires by email has been shown to be more convenient and to yield more
responses (Shannon & Bradshaw 2002). However, it was essential to investigate this method of distribution in case participants found the questionnaire difficult to complete electronically, in which case it would be administered by post. In addition, the pilot investigated whether participants would be able to complete the questionnaire without the researcher being present to explain questions or give additional information.

Potential participants were emailed to enquire about their willingness to take part in the pilot, which involved completing the questionnaire and informing the researcher of any comments or difficulties they had. Twelve individuals were sent questionnaires. Half were sent the questionnaire by email, and were required to complete it electronically and return it by email. The other half were sent it by post, were required to complete it on paper, and return it in a freepost envelope provided.

Completed questionnaires were received from 9 individuals. Of these, one attached emailed questionnaire was unable to be opened. This was thought to be due to the low quality, and probable incompatibility, of the individual’s home computer from which it had been completed and emailed. It was not thought that this would be a problem in the study however, as employees in large companies and organisations would be likely to work on computers with up-to-date software. One individual had pasted their completed questionnaire into the email which subsequently corrupted the formatting, making it invalid. It was decided that returning the questionnaire by Word document attachment would be strongly emphasised in the information given to participants with the questionnaire. Another individual returned their questionnaire too late to be of use in the pilot.

Therefore, six individuals successfully completed and returned their questionnaires, three by post and three by email.

All participants were then emailed again and asked the following questions:

- Did you find it easy to complete the questionnaire electronically/on paper?
- Would you have preferred to complete it electronically/on paper?
Did you find any of the questions too difficult or inappropriate to answer? If so, which ones?

Did you understand the information about the genetic test and your test results? If not, exactly what was difficult to understand?

Do you have any other comments about the questionnaire and the way it was sent and returned?

All individuals found the questionnaire easy and convenient. They also reported that the questions were understandable, the information relating to genetics was well explained and the questionnaire enjoyable to complete. The main difference between the two groups was that those who responded by email did so within a few days, whereas those who responded by post took at least a week.

Some participants were confused by some questions being repeated. For example, the anxiety scale and the risk perception statements are repeated before and after the test results are given in order to assess a change. One individual reported that he felt he was trying to be caught out and looked back at the questionnaire to check previous answers. A statement was therefore introduced at the beginning of the questionnaire which warned that some questions may be repeated and asked participants not to look back at any previous answers.

In addition to the pilot, the questionnaire was sent to an experienced researcher, with expertise in questionnaire design and hypothetical ‘vignette’ style studies and psychological constructs, particularly coping styles and risk perception.

It was advised that the risk perception statement was slightly altered from “What do you think your risk for developing heart disease is, compared with the average person?” to “What do you think your risk for developing heart disease is, compared with the average person the same age and gender as you?”

This latter comparative risk item is more appropriate as individuals may have otherwise compared themselves with those younger or older and responded with that comparison in mind. In addition, participants’ actual risk was unknown, in which case risk estimates could not be compared to absolute risk. Unrealistic optimism,
otherwise known as ‘optimistic bias’, is the belief that negative events are less likely
to happen to us, and positive events are more likely to happen to us, than to others.
However, without knowing actual risk, one cannot assess whether participants’ risk
estimates were optimistic or realistic. Comparative risk should be used in these
instances whereby comparative optimism can be detected rather than unrealistic
optimism (Weinstein & Klein 1996).

It was also suggested that the scenario was elaborated to make participants feel more
engaged in the situation. Research has shown that unrealistic risk perception can be
‘debiased’ by emphasising the situation as severe and blameworthy (McKenna &
Myers unpublished). In the present context, this would mean describing the severity
of heart disease, its symptoms and consequences, and causing the participant to feel
personally responsible for their risk, and for reducing it. The information about heart
disease and folic acid was changed to incorporate this.

**Estimation of sample size and power**

In order to achieve power of at least 0.8 with a significance level of 0.05, at least 80
participants were required in each condition (http://www.stat.uiowa.edu/~rlenth/
Power/ (with estimates of: SD x(j) = 0.3; Error SD = 3.0; and, detectable beta =
0.3)).

**6.2.3 Ethics approval**

The experimental questionnaire study invited healthy members of the general public
to participate in the study. Their consent was implicit in their application for a
questionnaire. No confidential, medical or personal data was requested from the
participants. Professor David Taylor consulted the Chair of the Camden and
Islington Ethics Committee, Ms Stephanie Ellis, who assured us that ethical approval
was not necessary.

**6.2.4 Participant recruitment**

Participants were recruited from five sources: Camden Council head office in
London, Boots plc head office in Nottingham, Hewlett Packard head office in
Camden Council head office, London

The Human Resources department at Camden Council was approached to enquire whether the research questionnaire could be sent to Council employees. It was agreed that an advert asking for participants would be emailed to Council staff via a 'Message of the Day' - a message which is emailed to approximately 2000 employees. An advert was designed which asked for volunteers to email the researcher if they were interested in completing the questionnaire. To increase the response rate, staff were told that all those replying to the message would be entered into a prize draw to win £100.

A total of 131 employees agreed to participate in the research and were sent the questionnaire together with a covering letter explaining the nature of the research and ensuring confidentiality and data protection of their responses. Allocation of participants to each condition was systematic, in that Test Result A was sent to the first participant who replied, Test Result B to the second participant, Test Result C to the third participant, Test Result A to the fourth participant, and so on. They were requested to complete the questionnaire electronically and return by email, although the option of receiving a hard copy and/or returning it by Freepost was also given. Fourteen were returned by post. Participants' consent to the research was implicit in their return of the completed questionnaire.

70 questionnaires had been received before a reminder email was sent 10-14 days after the first mailing. A further 27 completed questionnaires were returned after this. A total of 97 completed questionnaires were received back from a possible 131 questionnaires which were requested. A response rate cannot be calculated as it is not known how many staff saw the advert but did not respond.
September 2004 and was requested to be removed from the intranet on 18 October 2004. Entry into a £100 prize draw was offered in order to encourage responses.

A total of 142 individuals agreed to participate in the research and were sent the questionnaire together with a covering letter explaining the nature of the research and ensuring confidentiality and data protection of their responses. Allocation of participants to each condition was systematic, in that Test Result A was sent to the first participant who replied, Test Result B to the second participant, Test Result C to the third participant, Test Result A to the fourth participant, and so on. Participants' consent to the research was implicit in their return of the completed questionnaire.

They were requested to complete the questionnaire electronically and return by email, although the option of receiving a hard copy and/or returning it by Freepost was also given. Seventy five questionnaires had been received before a reminder email was sent to the remaining interested individuals 10-14 days after the first mailing. At least 24 completed questionnaires were returned in response to the reminder, an increase of 24%. A further 9 questionnaires were received by post anonymously.

A total of 108 completed questionnaires were returned, 93 by email and 15 by post. It is not possible to calculate a response rate as it was not known how many people accessed the webpage on which the questionnaire was advertised during the time the advert was live.

Hewlett Packard head office, Berkshire

The Human Resources department at Hewlett Packard agreed to distribute the questionnaire to some of its employees. All relevant documents were emailed to a contact in the HR department, who then circulated the questionnaire to a random sample of employees, ensuring random and equal distribution of the three different test results. In addition, questionnaires were given to staff in an ad hoc manner, for example, during staff training sessions.
Chapter Six – Experimental questionnaire study: Methods, results and discussion

A total of 52 completed questionnaires were received from Hewlett Packard employees. It was not possible to calculate a response rate as it was not known how many questionnaires were distributed.

School of Pharmacy, London

Ninety five non-academic staff at the School of Pharmacy (including technicians, maintenance, catering, finance and administrative) were sent questionnaires by internal post. The return of the questionnaires was also by internal post, ensuring anonymity and adding an additional mode of distribution to the study. Test results were randomly allocated to staff members.

A total of 30 responses had been received a month later when a reminder letter was sent to all. A further 9 completed questionnaires were subsequently received, increasing the response rate by 23%. A total of 39 completed questionnaires were received; a response rate of 41%.

The Association of the British Pharmaceutical Industry (ABPI), London

The Human Resources department at ABPI agreed for an employee to distribute 60 questionnaires to employees via email (20 of each condition, randomly allocated). A total of 7 completed questionnaires were received, a response rate of 11.6%, despite the offer of the £100 prize draw.

Miscellaneous office workers, Surrey

90 questionnaires (30 of each test result) were randomly distributed in pigeon holes in an office complex in South London. The complex housed various businesses such as a publishing company, a charity, an office supplies company, and an IT solutions business. A total of 31 completed questionnaires were returned by post; a response rate of 34%.

Unknown

It was not possible to determine the origin of 4 questionnaires received through the post.

Total sample

148
A total of 338 questionnaires were received (see Table 6.1).

Table 6.1: Origin of received questionnaires

<table>
<thead>
<tr>
<th>Number originally agreeing to participate in research</th>
<th>Number of completed questionnaires</th>
<th>% response</th>
</tr>
</thead>
<tbody>
<tr>
<td>Camden Council</td>
<td>131</td>
<td>97</td>
</tr>
<tr>
<td>School of Pharmacy</td>
<td>95</td>
<td>39</td>
</tr>
<tr>
<td>Boots Plc</td>
<td>142</td>
<td>108</td>
</tr>
<tr>
<td>Hewlett Packard (HP)</td>
<td>unknown</td>
<td>52</td>
</tr>
<tr>
<td>ABPI</td>
<td>60</td>
<td>7</td>
</tr>
<tr>
<td>Miscellaneous office workers</td>
<td>90</td>
<td>31</td>
</tr>
<tr>
<td>Unknown origin</td>
<td>4</td>
<td>TOTAL 338</td>
</tr>
</tbody>
</table>

6.2.5 Data entry and analysis

Quality control

Data was entered onto a database within SPSS v.13.0. After data had been entered it was checked for quality. Frequencies of all values were examined and extreme values searched for as they may indicate error in the database. No extreme values were found.

In addition, five percent (n = 17) of questionnaires from the whole sample were selected at random. The corresponding data entered onto SPSS was checked against the raw copies of these selected questionnaires. A total of three errors were found. The error rate of the whole sample was 0.01% (see Section 4.2.3).

6.3 Results

This chapter presents results from the questionnaire study. Specifically, section 6.3.1 will describe the characteristics of the sample in terms of demographics and section 6.3.2 will present reliability analyses of the measures and items used in the questionnaire. Section 6.3.3 presents results relating to the impact of genetic information on anxiety, perceived risk of heart disease and intention to change dietary behaviour. Sections 6.3.4, 6.3.5 and 6.3.6 describe findings relating to the
extent that psychological variables, coping style and information presentation respectively predict the impact of genetic health information. Finally, sections 6.3.7 and 6.3.8 respectively present results concerning the extent that individuals were concerned about the use of genetic information and how well they understood the information they were given.

6.3.1 Sample demographics

6.3.1.1 Age

Table 6.2: Age group frequency and percentage in the study sample and Census 2001 data.

<table>
<thead>
<tr>
<th>Missing n=3 (0.9%)</th>
<th>FREQUENCY AND PERCENTAGE (%) OF CASES</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>18-20</td>
</tr>
<tr>
<td>Sample</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>1.2%</td>
</tr>
<tr>
<td>Census 2001*</td>
<td>15-19 yrs</td>
</tr>
<tr>
<td>9.6%</td>
<td>9.2%</td>
</tr>
</tbody>
</table>

Table 6.2 shows that the majority of study participants were in the age groups 31 to 35 years (19%) and 36 to 40 years (18%). Sixteen percent were in the age group 51+ years. The proportion in this latter age category is likely to be slightly higher than others due to it capturing participants from 51 to retirement age (60 years old for women, 65 for men). Thirteen percent were in age groups 41 to 45 and 46 to 50, and 11% in the age group 26 to 30 years. Younger participants were fewer, with 10% being aged 21 to 25 and only 1% aged 18 to 20 years old. This probably reflects a fairly accurate percentage of this young age group in a working environment as many 18 to 25 year olds may be studying rather than working.

Table 6.2 also presents data taken for the Census 2001 to compare the study sample demographics to that of UK population. The Census 2001 gives age ranges in 5-year blocks starting from 0-4 years to 85-89 years, then 90 and over as the last category. In order to compare more accurately, only the working age population (15 to 64
years of age) is presented. Percentages were calculated using the total UK population between 15 and 64 years of age.

The higher proportion in the Census sample of the lowest age range could be attributed to the Census block being of 5 years (15 to 19 years of age) as opposed to the study sample being a 3-year block (18 to 20 year olds), in addition to a low proportion of 18 to 20 year olds being employed in work, as mentioned earlier. Similarly, the higher proportion in the Census sample of the highest age bracket could be attributed to the Census block being of 15 years (50 to 64 years of age) as opposed to the study sample being 51+, where many people, especially women, retire at 60 years of age.

Figure 6.2: Age distribution of the study sample compared to the population in England.

Figure 6.2 displays the study sample and the Census sample visually, indicating similar distributions of ages, except for those mentioned above, and much higher proportions of 31-35 year olds and 36-40 year olds in the study sample.

### 6.3.1.2 Gender

The majority of participants were female (68.3%, n = 231). It is possible that, in general, more females than males work in office environments and therefore more women may have completed the questionnaire. In addition, women tend to be more
interested in health issues than men which could have resulted in this gender bias (Green & Pope 1999).

6.3.1.3 Marital status

Table 6.3: Distribution of marital status within study sample and from Census 2001.

<table>
<thead>
<tr>
<th>Missing n= 3 (0.9%)</th>
<th>SINGLE</th>
<th>MARRIED</th>
<th>CO-HABITING</th>
<th>DIVORCED</th>
<th>WIDOWED</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sample</td>
<td>n</td>
<td>%</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>98</td>
<td>29%</td>
<td>157</td>
<td>64</td>
<td>15</td>
<td>1</td>
</tr>
<tr>
<td>157</td>
<td>46.4%</td>
<td>64</td>
<td>15</td>
<td>1</td>
<td>0.3%</td>
</tr>
<tr>
<td>64</td>
<td>18.9%</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4.4%</td>
<td>4.4%</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1</td>
<td>1.6%</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

2003 estimates from Census 2001* (England & Wales, aged 20 to 64)

<table>
<thead>
<tr>
<th>Missing n= 3 (0.9%)</th>
<th>SINGLE</th>
<th>MARRIED</th>
<th>CO-HABITING</th>
<th>DIVORCED</th>
<th>WIDOWED</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sample</td>
<td>n</td>
<td>%</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>152</td>
<td>32.5%</td>
<td>55.7%</td>
<td>No information</td>
<td>10.2%</td>
<td>1.6%</td>
</tr>
</tbody>
</table>


The majority of participants were married (46.4%) with 29% being single and 18.9% cohabiting. Divorced and widowed individuals made up 4.4% and 0.3% of the sample respectively. To compare with the 2003 estimates for England and Wales (based on the Census 2001) more accurately, percentages were calculated from ages 20 to 64. The proportions of marital status in the study sample and from the Census are fairly similar (see Table 6.3). The Census estimate has a slightly larger proportion of widowed individuals, and a much larger proportion of divorced individuals. Unfortunately, there is no Census data on co-habitation.
6.3.1.4 Ethnicity

Table 6.4: Distribution of ethnicity in sample and Census 2001 data.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Sample N</th>
<th>Census 2001 (England)</th>
</tr>
</thead>
<tbody>
<tr>
<td>White</td>
<td>295</td>
<td>92%</td>
</tr>
<tr>
<td>Indian</td>
<td>14</td>
<td>1.8%</td>
</tr>
<tr>
<td>Pakistani/ Bangladeshi</td>
<td>2</td>
<td>1.8%</td>
</tr>
<tr>
<td>Black Caribbean</td>
<td>7</td>
<td>1%</td>
</tr>
<tr>
<td>Black African</td>
<td>3</td>
<td>0.8%</td>
</tr>
<tr>
<td>Chinese</td>
<td>5</td>
<td>0.4%</td>
</tr>
<tr>
<td>Mixed</td>
<td>7</td>
<td>1.2%</td>
</tr>
<tr>
<td>White Irish</td>
<td>1</td>
<td>Unknown</td>
</tr>
<tr>
<td>Other</td>
<td>1</td>
<td>1%</td>
</tr>
</tbody>
</table>

The vast majority of the sample (87.3%) were white. The next largest ethnic group in the whole sample was Indian (4.1%), followed by black Caribbean (2.1%) and mixed (2.1%). Chinese, black African, Pakistani/Bangladeshi, white Irish and other ethnicity made up 1.5%, 0.9%, 0.6%, 0.3% and 0.3% respectively. Table 6.4 shows the sample data alongside data from the 2001 Census. The Census data reports that in England in 2001, 92% of the population were white, with between 0.8% and 1.8% belonging to the other ethnic categories.

Due to low numbers in all ethnicity categories other than white, non-white categories were collapsed into one and comprised 11.8% of the study sample.
6.3.1.5 Education

Table 6.5: Distribution of highest qualification in study sample and UK population.

<table>
<thead>
<tr>
<th>Missing n = 7 (2.1%)</th>
<th>Study sample</th>
<th>Government statistics Winter 2003 (UK)*</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>(n) % of sample</td>
<td></td>
</tr>
<tr>
<td>None</td>
<td>(13) 3.8%</td>
<td>14.4%</td>
</tr>
<tr>
<td>GCSE</td>
<td>(54) 16.0%</td>
<td>21.9%</td>
</tr>
<tr>
<td>A level</td>
<td>(38) 11.2%</td>
<td>24.1%</td>
</tr>
<tr>
<td>Higher education</td>
<td>(56) 16.6%</td>
<td>8.6%</td>
</tr>
<tr>
<td>Degree or equivalent</td>
<td>(133) 39.3%</td>
<td>17.1%</td>
</tr>
<tr>
<td>Professional qualification</td>
<td>(3) 0.9%</td>
<td>Information not available</td>
</tr>
<tr>
<td>Postgraduate (PG Cert, PG Dip, PGCE)</td>
<td>(13) 3.9%</td>
<td></td>
</tr>
<tr>
<td>Masters degree</td>
<td>(18) 5.3%</td>
<td></td>
</tr>
<tr>
<td>DPhil</td>
<td>(1) 0.3%</td>
<td></td>
</tr>
<tr>
<td>Doctorate</td>
<td>(2) 0.6%</td>
<td></td>
</tr>
<tr>
<td>Other qualifications</td>
<td></td>
<td>13.2%</td>
</tr>
</tbody>
</table>


Table 6.5 shows that the majority of the sample reached degree level or equivalent in their education (39.3%). 16.6% had higher education qualifications, 16% had GCSE’s and 11.2% had A levels. A postgraduate qualification was the highest level of education for 3.9% of the sample and a Masters degree for 5.3%. Four percent reported no educational qualifications.

Table 6.5 also presents UK statistics for highest educational level. It shows that, compared with the rest of the UK, the study sample is particularly well educated. Only 17.1% of the UK population holds a degree or equivalent as their highest qualification compared with 39.3% of the study sample. The comparison can be seen more clearly in Figure 6.3 below (note that information is not available for national postgraduate figures). This could be a reflection of the study recruitment process in which many participants were made aware of the study via Intranet, which requires access to a computer. Individuals in jobs which do not require a computer were essentially excluded from the study and these positions may have been occupied by less educated individuals.
Figure 6.3: Distribution of highest educational qualification in whole sample and in UK.

The categories of ‘higher education’ and ‘degree or equivalent’ were eventually collapsed into one category of ‘Higher education’, which comprised 55.9% of the study sample. Cases reporting their highest qualification as PG Certificate, PG Diploma, PGCE, Professional qualification, Masters degree, DPhil or Doctorate were collapsed into one category labelled ‘Postgraduate’ which made up 11% of the study sample.

In conclusion, the study sample was largely female, married, White and well educated.

6.3.2 Reliability analysis of scales and measures and frequency distribution of scores

This section will present results from the reliability analyses performed on the measures used in the questionnaire. Section 4.2.3 described reliability analysis in more detail.
Chapter Six — Experimental questionnaire study: Methods, results and discussion

6.3.2.1 Multidimensional Health Locus of Control (MHLC – Form A): Wallston, Wallston & DeVellis (1978)

MHLC-Internal subscale (MHLC-I)

Within the MHLC scale items 1, 6, 8, 12, 13 and 17 were attributed to measuring the Internal subscale. Each item was scored between 1 and 5. The six items were summed to make a score, with a possible range of 6 to 30 and a midpoint of 18. No items needed to be reversed before summing.

### Table 6.6: Item-Total statistics and Cronbach’s alpha for MHLC-I

<table>
<thead>
<tr>
<th>Item</th>
<th>Corrected item-total correlation (r)</th>
<th>Cronbach’s alpha (α)</th>
<th>Cronbach’s alpha if item deleted (α)</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Item 1) If I get ill, it is my own behaviour which determines how soon I get well again</td>
<td>0.35</td>
<td>0.71</td>
<td>0.70</td>
</tr>
<tr>
<td>(Item 6) I am in control of my health</td>
<td>0.46</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(Item 8) When I get ill, I am to blame</td>
<td>0.24</td>
<td>0.73</td>
<td></td>
</tr>
<tr>
<td>(Item 12) The main thing which affects my health is what I myself do</td>
<td>0.56</td>
<td></td>
<td>0.63</td>
</tr>
<tr>
<td>(Item 13) If I take care of myself, I can avoid illness</td>
<td>0.49</td>
<td>0.65</td>
<td></td>
</tr>
<tr>
<td>(Item 17) If I take the right actions, I can stay healthy</td>
<td>0.59</td>
<td></td>
<td>0.63</td>
</tr>
</tbody>
</table>

Table 6.6 shows that most items performed well within the subscale. This is indicated by the corrected item-total correlations being at least \( r = 0.3 \). However, Item 8 (“When I get ill, I am to blame”) seems to perform less well, shown by a low item-total correlation of \( r = 0.24 \). This suggests that participants responded differently to it, tending to agree with it less compared to the other statements.

Cronbach’s alpha is satisfactory at \( \alpha = 0.71 \). Other research has reported \( \alpha = 0.75 \) (Marshall, Collins & Crooks 1990). However, Item 8 is the only item to increase the alpha value if deleted, further suggesting its poor performance within the whole scale.
The distribution of scores remained normal when Item 8 was removed (see Figure 6.4) therefore it was not included in subsequent analyses. With five items the midpoint of the scale became 15, indicated by the X axis reference line. The distribution of scores is clearly negatively skewed indicating that the majority of the sample scored highly on this scale.

**MHLC-Chance subscale (MHLC-C)**

Within the MHLC scale items 2, 4, 9, 11, 15 and 16 were attributed to measuring the Chance subscale. Each item was scored between 1 and 5. The six items were summed to make a score, which could range from 6 to 30 with a midpoint of 18. No items needed to be reversed before summing.
Table 6.7: Item-Total statistics and Cronbach’s alpha for the MHLC-C

<table>
<thead>
<tr>
<th>Item</th>
<th>Corrected item-total correlation (r)</th>
<th>Cronbach’s alpha (α)</th>
<th>Cronbach’s alpha if item deleted (α)</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Item 2) No matter what I do, if I am going to get ill, I will get ill</td>
<td>0.53</td>
<td>0.73</td>
<td>0.67</td>
</tr>
<tr>
<td>(Item 4) Most things that affect my health happen to me by accident</td>
<td>0.40</td>
<td></td>
<td>0.71</td>
</tr>
<tr>
<td>(Item 9) Luck plays a big part in determining how soon I will recover from an illness</td>
<td>0.56</td>
<td>0.73</td>
<td>0.66</td>
</tr>
<tr>
<td>(Item 11) My good health is largely a matter of good fortune</td>
<td>0.50</td>
<td></td>
<td>0.68</td>
</tr>
<tr>
<td>(Item 15) No matter what I do, I’m likely to get ill</td>
<td>0.37</td>
<td>0.73</td>
<td>0.71</td>
</tr>
<tr>
<td>(Item 16) If it’s meant to be, I will stay healthy</td>
<td>0.41</td>
<td></td>
<td>0.70</td>
</tr>
</tbody>
</table>

Table 6.7 shows that most items performed well within the scale. Cronbach’s alpha is satisfactory at α = 0.73. A previous study of outpatients reported α = 0.63 (Marshall, Collins & Crooks 1990). None of the items increase the alpha value if deleted.

Figure 6.5: Distribution of scores on the MHLC-C subscale

When checking for normal distribution, Figure 6.5 suggested a possible bimodal score distribution. However, other indications suggest otherwise. The normality curve indicates that the scores are relatively normally distributed. In addition, the Q-Q plot (see Figure 6.6 below) shows that the observed values (individual points) do
not seem to deviate substantially from the expected values (the straight line). As a result, it was decided that the MHLC-C scores could be treated as normally distributed.

**Figure 6.6: Normal Q-Q plot for MHLC-C**

MHLC-Powerful others subscale (MHLC-P)

Within the MHLC scale items 3, 5, 7, 10, 14 and 18 were attributed to measuring the Powerful Others subscale. Each item was scored between 1 and 5. The six items were summed to make a score, which could range from 6 to 30 with a midpoint of 18. No items needed to be reversed before summing.

**Table 6.8: Item-Total statistics and Cronbach’s alpha for MHLC-P**

<table>
<thead>
<tr>
<th>Item</th>
<th>Corrected item-total correlation (r)</th>
<th>Cronbach’s alpha ((\alpha))</th>
<th>Cronbach’s alpha if item deleted ((\alpha))</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Item 3) Having regular contact with my doctor is the best way for me to avoid illness</td>
<td>0.53</td>
<td>0.73</td>
<td>0.67</td>
</tr>
<tr>
<td>(Item 5) Whenever I don’t feel well, I should consult a medically trained professional</td>
<td>0.59</td>
<td></td>
<td>0.65</td>
</tr>
<tr>
<td>(Item 7) My family has a lot to do with my becoming ill or staying healthy</td>
<td>0.11</td>
<td></td>
<td>0.79</td>
</tr>
<tr>
<td>(Item 10) Health professionals control my health</td>
<td>0.43</td>
<td></td>
<td>0.70</td>
</tr>
<tr>
<td>(Item 14) Whenever I recover from an illness, it’s usually because other people (for example, doctors, nurses, family, friends) have been taking good care of me</td>
<td>0.56</td>
<td></td>
<td>0.66</td>
</tr>
<tr>
<td>(Item 18) Regarding my health, I can only do what my doctor tells me to do</td>
<td>0.64</td>
<td></td>
<td>0.63</td>
</tr>
</tbody>
</table>
Table 6.8 shows that the majority of items performed well within the scale. However, Item 7 ("My family has a lot to do with my becoming ill or staying healthy") seems to perform less well, indicated by a low item-total correlation of $r = 0.111$. This suggests that participants respond differently to it. Cronbach’s alpha for the MHLC-P is satisfactory at $\alpha = 0.73$. However, Item 7 is the only item to increase the alpha value if deleted, further suggesting its poor performance within the whole scale.

Marshall, Collins & Crooks (1990) reported a Cronbach’s alpha of $\alpha = 0.64$ for this subscale. They also conducted a factor analysis of the MHLC which revealed a three-factor structure with all but one item significantly loading only on the factors which corresponded to their a priori subscales. The item which did not load onto the appropriate subscale was equivalent to Item 7. Subsequent studies have also found this (Bonetti et al 2001). It is possible that family influences over health are not as strong as they once were. Many individuals are now separated from their families, perhaps living in different towns, and do not have as much contact as in the 1970s when the scale was developed. This item therefore may be outdated.

Figure 6.7: Distribution of scores on the MHLC-P, with Item 7 removed.

The distribution of scores remained normal when Item 7 was removed (see Figure 6.7) and therefore subsequent analyses using the MHLC-P did not include Item 7.
With five items the midpoint of the scale became 15, indicated by the X axis reference line. The distribution of scores is clearly positively skewed indicating that the majority of the sample scored low on this scale.

The article which cites the development of the MHLC scale (Wallston, Wallston & DeVellis 1978) reports Cronbach’s alpha coefficients of 0.77, 0.67 and 0.75 for the Internal, Powerful Others and Chance subscales on Form A respectively.

Several studies have reported Cronbach’s alpha to be higher on the Internal subscale than the Powerful Others or Chance subscales, as has been reported here. For example, Cooper & Fraboni (1990) reported alpha’s of 0.87, 0.68 and 0.76 and respectively on MHLC Form A.

Several studies have supported the three-factor structure of the MHLC (Bonetti et al 2001).

6.3.2.2 The Health Value scale: Lau, Hartman & Ware (1986)

The Health Value Scale is a 4-item scale. Items 1 and 3 were reverse coded and summed with Items 2 and 4 to produce a total score. A five-point response format was used making the possible range of scores between 4 and 20, with a midpoint of 12.

Table 6.9: Item-Total statistics and Cronbach’s alpha for the Health Value scale

<table>
<thead>
<tr>
<th>Item</th>
<th>Corrected item-total correlation (r)</th>
<th>Cronbach’s alpha (α)</th>
<th>Cronbach’s alpha if item deleted (α)</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Item 1) There is nothing more important than good health</td>
<td>0.38</td>
<td>0.68</td>
<td>0.66</td>
</tr>
<tr>
<td>(Item 2) Good health is only of minor importance in a happy life</td>
<td>0.53</td>
<td></td>
<td>0.56</td>
</tr>
<tr>
<td>(Item 3) If you don’t have your health you don’t have anything</td>
<td>0.55</td>
<td></td>
<td>0.55</td>
</tr>
<tr>
<td>(Item 4) There are many things I care about more than my health</td>
<td>0.39</td>
<td></td>
<td>0.66</td>
</tr>
</tbody>
</table>
Table 6.9 shows that all four items performed well in the scale. Cronbach's alpha is satisfactory at $\alpha = 0.68$. This corresponds well with other research using the Health Value Scale. Lau, Hartman & Ware (1986) reported Cronbach's alphas of 0.67 in a study of undergraduates and 0.63 in a study of parents of undergraduates. None of the items increase the alpha value if deleted, further showing that all items performed well within the scale. Figure 6.8 confirms that the distribution of scores is normal.

6.3.2.3 The short-form state scale of the Spielberger State-Trait Anxiety Inventory (STAI): Marteau & Bekker (1992)

The short-form STAI consists of 6 items scored on a 4-point scale. Items 1, 4 and 5 are reverse coded and scores are summed with scores from items 2, 3 and 6 to give a total score with a possible range of 6 to 24 and a midpoint of 15. The scale was presented at two time points within the questionnaire. Reliability analyses from both time points are shown here.
Table 6.10: Item-Total statistics and Cronbach’s alpha for the STAI.

<table>
<thead>
<tr>
<th>Item</th>
<th>Time 1</th>
<th>Time 2</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Corrected item-total correlation (r)</td>
<td>Cronbach’s alpha (α)</td>
</tr>
<tr>
<td>(Item 1)</td>
<td>0.65</td>
<td>0.75</td>
</tr>
<tr>
<td>Calm</td>
<td>0.80</td>
<td></td>
</tr>
<tr>
<td>(Item 2)</td>
<td>0.63</td>
<td>0.76</td>
</tr>
<tr>
<td>Tense</td>
<td>0.79</td>
<td>0.55</td>
</tr>
<tr>
<td>(Item 3)</td>
<td>0.51</td>
<td>0.55</td>
</tr>
<tr>
<td>Upset</td>
<td>0.69</td>
<td></td>
</tr>
<tr>
<td>(Item 4)</td>
<td>0.61</td>
<td>0.79</td>
</tr>
<tr>
<td>Relaxed</td>
<td>0.79</td>
<td>0.60</td>
</tr>
<tr>
<td>(Item 5)</td>
<td>0.50</td>
<td>0.79</td>
</tr>
<tr>
<td>Content</td>
<td>0.50</td>
<td>0.79</td>
</tr>
<tr>
<td>(Item 6)</td>
<td>0.50</td>
<td>0.79</td>
</tr>
<tr>
<td>Worried</td>
<td>0.50</td>
<td>0.79</td>
</tr>
</tbody>
</table>

Figure 6.9: Distribution of scores on the short-form STAI (Time 1)
All items within the scale seem to perform well at both time points (Table 6.10). Cronbach’s alpha is also satisfactory at both time points (α = 0.80 at Time 1; α = 0.85 at Time 2). This corresponds well with the original validation of the scale which reported a Cronbach’s alpha of α = 0.82 (Marteau & Bekker 1992). None of the items increase the alpha value if deleted, further showing that all items performed well within the scale.

Figures 6.9 and 6.10 show that the distribution of scores at both time points can be considered normal. The X axis references lines at the mid-point of 15 indicate that the sample tended to score low on the scale at both time points.

For some analyses it was necessary to dichotomise the STAI scores into high or low anxiety. Cases were split at the score of 15, the mid-point of the scale. Because the majority of cases were below the mid-point at both time points it was decided that cases with a score of 15 would be categorised as high, in order to distribute the cases into the two categories as evenly as possible.

Although it is common to dichotomise continuous variables, some critics do not believe that it is good practice. For example, MacCallum, Zhang, Preacher & Rucker (2002) criticise dichotomisation except in circumstances when it is clear that two
distinct groups exist, or when the distribution of a variable is skewed. At any other times the research can be compromised due to loss of statistical power and possibly erroneous results. For these reasons care is taken when discussing results involving this measure.

6.3.2.4 The Nutrition Self-Efficacy scale: Schwarzer & Renner (2000)

The Nutrition Self-Efficacy Scale consists of five items scored on a 4-point scale. The items were summed to give a total score with a possible range of 5 to 20 and a midpoint of 12.5.

Table 6.11: Item-Total statistics and Cronbach’s alpha for the Nutrition Self-Efficacy scale.

<table>
<thead>
<tr>
<th>Item</th>
<th>Corrected item-total correlation (r)</th>
<th>Cronbach's alpha (α)</th>
<th>Cronbach's alpha if item deleted (α)</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Item 1) Even if you need a long time to develop the necessary routines</td>
<td>0.82</td>
<td>0.95</td>
<td>0.94</td>
</tr>
<tr>
<td>(Item 2) Even if you have to try several times until it works</td>
<td>0.88</td>
<td>0.93</td>
<td>0.93</td>
</tr>
<tr>
<td>(Item 3) Even if you have to rethink your entire way of nutrition</td>
<td>0.89</td>
<td>0.93</td>
<td>0.93</td>
</tr>
<tr>
<td>(Item 4) Even if you do not receive a great deal of support from others when making your first attempts</td>
<td>0.85</td>
<td>0.93</td>
<td>0.93</td>
</tr>
<tr>
<td>(Item 5) Even if you have to make a detailed plan</td>
<td>0.84</td>
<td>0.94</td>
<td>0.94</td>
</tr>
</tbody>
</table>

Figure 6.11: Distribution of scores on the Nutrition Self-Efficacy Scale

![Distribution of scores on the Nutrition Self-Efficacy Scale](image)
Chapter Six – Experimental questionnaire study: Methods, results and discussion

No items within the scale performed poorly, indicated by the inter-item correlations of at least 0.8 (Table 6.11). Cronbach’s alpha is also very high at $\alpha = 0.95$; much higher than a previous report of $\alpha = 0.87$ (Schwarzer & Renner 2000). Although this could be seen as indicating a very reliable scale, it actually suggests that there was little variability in the responses to the items within the scale. In other words, one item would be sufficient to obtain the same score as all the items. Mean scores for all items was >3, indicating that participants scored at the high end of the response scale. Figure 6.11 confirms this by showing non-normally distributed scores with high peaks at 15 and 20. Together, this suggests that participants tended to score the same on all items, on the third or fourth point of the scale.

Due to the non-normally distributed scores, it was decided that cases would be recoded as either high or low scorers on the scale. Cases were split at the mid-point of the scale (12.5) so that cases scoring less than or equal to twelve were classified as low scorers, and cases scoring more than twelve were classified as high scorers.

As mentioned earlier, dichotomisation of continuous variables has its limitations. However, it was decided that in this particular case dichotomisation of the scale would at least allow its inclusion in some analysis of the data, and that care would be taken when discussing results.

6.3.2.5 Miller Behavioral Style Scale (MBSS): Miller (1987)

The MBSS consists of four hypothetical scenarios. Each scenario is followed by eight statements detailing possible response behaviours. Participants are asked to mark which behaviours they would be likely to perform in response to each given scenario.

Four out of eight possible behaviours are examples of ‘monitor’ coping responses, and the other four are examples of ‘blunter’ responses. Total monitor and total blunter responses are calculated, each with a possible range of 0 to 16. The median split can be used to characterise individuals as high or low monitors or blunters. Note that it is also possible to calculate a ‘summary score’ by subtracting the total blunter
score from the total monitor score. However, using two separate scores may be more appropriate as previous research has suggested that monitor and blunter concepts are two separate dimensions (Myers & Derakshan 2000). For example, a high-scoring monitor is not necessarily a low-scoring blunter.

Miller suggests dividing cases into high or low monitors or blunters by dividing at the median. The median for the total monitoring scores was 7, and the median for the total blunting scores was 4. Because large numbers of cases scored exactly the median value, the mean was used to decide in which half the median scoring cases would be allocated. The mean of the monitoring scores was 7.27, indicating that cases scoring 7 should be classed as 'low monitors'. The mean of the blunting scores was 3.94, indicating that cases scoring 4 should be classed as 'high blunters'. Indeed, allocating median scoring cases in this way allowed the categories to become fairly even, with 51.8% of cases in 'low monitor' category, and 45% of cases in the 'high blunter' category, compared to 40.5% and 64.5% if median cases had been allocated in the other category.

Miller has not published any description of a method of reliability analysis of the MBSS. Cronbach's alpha coefficients from some experiments using the MBSS are reported in Miller (1987): 0.79 and 0.75 for the monitoring items, and 0.69 and 0.67 for the blunting items.

Muris & Schouten (1994) used a 5-point version of the MBSS, where individuals score each item from 1 to 5 ("not at all" to "very much") on how well that behaviour describes what they would do in that situation. The correlation between monitoring and blunting scores was $r = 0.11$. Cronbach's alpha for monitoring items was 0.79 and for blunting items was 0.69.

In another study, Muris et al (1994) examined the factor structure of the MBSS which revealed two-factors, both on the scale as a whole and when carried out on situation-specific items. Except for three items which did not load onto either factor, monitoring items loaded onto Factor 1 and blunting items loaded onto Factor 2.
The use of Guttman scoring, or ‘scalogram’ analysis was investigated as a method of testing the reliability of the MBSS. This technique is used on scales where participants endorse a category. However, items on a Guttman scale are cumulative, that is, if one ticked the last category, one would also have ticked all the previous categories. This meant that it could not be used on the MBSS as it is not a cumulative scale, that is, categories are not dependent on the responses to any other category. Factor analysis of the MBSS was considered but rejected as responses to the scale are binary (factor analysis is a correlational technique which requires linear scores).

It was therefore assumed that the MBSS was a reliable instrument due to previous citations of reliability.

6.3.2.6 Risk perception

Risk perception was scored on a visual analogue scale measuring 10cm long on the questionnaire with Very Low at one end, Very High at the other, and Average indicated in the middle. It was measured with a ruler so that scores could range from 0 to 100 (millimetres). The measure was taken at two time points.

Figure 6.12: Distribution of risk perception scores at Time 1
Figure 6.13: Distribution of risk perception scores at Time 2

Figures 6.12 and 6.13 indicate that the distribution of scores is normal.

6.3.2.7 Intention to change diet

As with risk perception, intention to change diet was scored on a visual analogue scale measuring 10cm long on the questionnaire with *Very Unlikely* at one end and *Very Likely* at the other. It was measured with a ruler so that scores could range from 0 to 100 (millimetres).

Figure 6.14: Distribution and normality curve of Intention to Change Diet scores.
The distribution of the Intention to Change Diet score was non-normal (Figure 6.14). In this case it was necessary to use nonparametric statistical tests for analysis involving this measure.

6.3.2.8 Health beliefs
Beliefs about the extent to which individuals believed that diet, genes or lifestyle in general influenced health was measured using a five-point scale, giving a possible range of 1 to 5 for each item.

Figure 6.15: Score distribution of diet-related health beliefs at Time 1.

Figure 6.16: Score distribution of lifestyle-related health beliefs at Time 1.

Figure 6.17: Score distribution of gene-related health beliefs at Time 1.
Figure 6.18: Score distribution of diet-related health beliefs at Time 2.

Figure 6.19: Score distribution of lifestyle-related health beliefs at Time 2.

Figure 6.20: Score distribution of gene-related health beliefs at Time 2.

Histogram charts (Figures 6.15 to 6.20) indicate that scores were negatively skewed but normally distributed at both time points.
6.3.2.9 **Self-rated health**

Self-rated health was measured using a single item on a five-point scale. Figure 6.21 shows that the score distribution was slightly negatively skewed, with a mean score of 3.76, indicating that most people rated their health as average to good.

**Figure 6.21: Score distribution of self-rated health measure.**

6.3.2.10 **Dietary information: Fruit and vegetable intake and sugar and fat intake**

Intake of fruit and vegetables was measured using a four-point scale of ‘less than 2’, ‘2 or 3’, ‘4’ or ‘at least 5’. Sugar and fat intake was measured using a five-point scale from ‘hardly ever’ to ‘every day’. These measures were taken as a general indication of diet.

6.3.2.11 **Physical activity level**

Level of physical activity was scored on a visual analogue scale measuring 10 cm long on the questionnaire with *Not at all active* at one end and *Very active* at the other. It was measured with a ruler so that scores could range from 0 to 100 (millimetres).

**Figure 6.22** suggests that the distribution of the score was negatively skewed.
6.3.3 What is the impact of genetic health risk information on anxiety, perceived risk of heart disease and intention to change diet?

In order to assess the impact of genetic health risk information, the sample was divided into those who were told they had the MTHFR gene variation (Gene group) and those who were told that they did not (No Gene group).

6.3.3.1 Anxiety
A One-Way Analysis of Variance (ANOVA) with a post-hoc Bonferroni test was conducted on anxiety levels in the three test conditions. In Condition C (gene-negative) anxiety levels after receipt of risk information differed significantly from both Condition A (gene-positive, gain framed information) and Condition B (gene-positive, loss framed information) (means = 10.01, 13.69 and 13.96 respectively; F = 48.17, p <0.001). Condition A did not differ significantly from Condition B.

In addition, Conditions A and B were grouped together to form the Gene group (n = 195), and anxiety scores before and after receipt of genetic information were compared with the No Gene group (n = 101) using paired t-tests.
Figure 6.23 shows that at baseline, anxiety scores were similar in the Gene and No Gene groups (mean scores of 11.61 and 11.07 respectively; \( t = 0.77, p = 0.441 \)). Anxiety scores after receipt of genetic information (mean = 13.84) were significantly higher than baseline scores in the Gene group (\( t = -8.49, p<0.0001 \)). In the No Gene group, anxiety scores after receipt of information (mean = 10.0) were significantly lower than baseline scores (\( t = 3.38, p<.0001 \)). Additionally, anxiety levels after receipt of genetic information in the Gene group were significantly higher than in the No Gene group (\( t = 10.78, p<0.0001 \)).

6.3.3.2 Perceived risk
A One-Way Analysis of Variance (ANOVA) with a post-hoc Bonferroni test was conducted on risk perception scores in the three test conditions. In Condition C (gene-negative) perceived risk of heart disease after receipt of risk information
differed significantly from both Condition A (gene-positive, gain framed information) and Condition B (gene-positive, loss framed information) (means = 39.41, 60.63 and 59.48 respectively; F = 36.73, p < 0.001). Condition A did not differ significantly from Condition B.

In addition, Conditions A and B were grouped together to form the Gene group (n = 212), and perceived risk of heart disease before and after receipt of genetic information were compared with the No Gene group (n = 110) using paired t-tests.

Figure 6.24: Risk perception scores before and after receipt of test results, in Gene and No Gene groups

Figure 6.24 shows that at baseline, risk perception scores were similar in the Gene and No Gene groups (mean scores of 43.29 and 43.71 respectively; t = -0.46, p = 0.643). Perceived risk after receipt of genetic information (mean = 60.10) was significantly higher than at baseline in the Gene group (t = -10.82, p < 0.0001). In the No Gene group, perceived risk after receipt of information (mean = 39.25) was
significantly lower than at baseline (t = 2.81, p = 0.006). Additionally, risk perception after receipt of genetic information in the Gene group was significantly higher than in the No Gene group (t = 8.57, p<0.0001).

6.3.3.3 Intention to change diet
Due to the non-normal distribution of the intention to change diet score the nonparametric Kruskall-Wallis test was conducted to compare the three test conditions. Intention to change diet was significantly different between conditions A, B and C with mean ranks of 172.49, 196.31 and 119.17 respectively (p<0.001).

Participants in conditions A and B were grouped together to form the Gene (n = 213) group and were compared with the No Gene group (n = 110) using the nonparametric Mann-Whitney U test. The Gene group had a significantly higher intention to change diet score than the No Gene group (Median = 81 and 57.5 respectively; U=7004, p<0.0001).

6.3.3.4 Fatalism and false reassurance
Spearman’s correlations show that, in the Gene group, risk perception after receipt of test results and intention to change behaviour are not significantly correlated (rho = 0.07, p = 0.26). This suggests that participants who received ‘positive’ genetic information did not become fatalistic.

Moreover, risk perception after receipt of test results and intention to change behaviour in the No Gene group was not significantly correlated (rho = 0.01, p = 0.92), indicating that false reassurance in those given ‘negative’ test results was not apparent in this sample.
6.3.4 **Do demographic factors or health indicators predict reactions to the test results?**

The data was further analysed with the aim of exploring whether demographic variables or health indicators affected the impact of genetic risk information on anxiety, perceived risk of heart disease or intention to change diet.

### 6.3.4.1 Anxiety

The following analysis was carried out with the aim of exploring whether anxiety levels differed according to demographic variables or health indicators.

**Age**

The 18-20 year old age group was omitted from analyses as it contained only one case. A One-Way Analysis of Variance (ANOVA) with a post-hoc Bonferroni test was conducted on anxiety levels in age groups. In the Gene group anxiety levels after receipt of risk information of both the 21 to 25 year olds and the 46 to 50 year olds differed significantly from the over 51s (means = 15.22, 14.64 and 11.67 respectively; F = 2.497, p = 0.018). No differences were found between age groups in the No Gene group.

**Gender**

A General Linear Model (GLM) using repeated measures was used to investigate the interactions between gender and gene status on anxiety levels.
Table 6.12: Gender differences in anxiety at baseline and after test results.

<table>
<thead>
<tr>
<th></th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Within-Subjects</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Repeat</td>
<td>33.36</td>
<td>1</td>
<td>33.36</td>
<td>5.38</td>
<td>0.02</td>
</tr>
<tr>
<td>Repeat * Gender</td>
<td>1.07</td>
<td>1</td>
<td>1.07</td>
<td>0.17</td>
<td>0.68</td>
</tr>
<tr>
<td>Repeat * Gene</td>
<td>276.85</td>
<td>1</td>
<td>276.85</td>
<td>44.61</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gene * Gender</td>
<td>5.31</td>
<td>1</td>
<td>5.31</td>
<td>0.86</td>
<td>0.36</td>
</tr>
<tr>
<td>Error (repeat)</td>
<td>1799.80</td>
<td>290</td>
<td>6.21</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Between-Subjects</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Intercept</td>
<td>61145.23</td>
<td>1</td>
<td>61145.23</td>
<td>4447.77</td>
<td>0.00</td>
</tr>
<tr>
<td>Gender</td>
<td>0.19</td>
<td>1</td>
<td>0.19</td>
<td>0.01</td>
<td>0.91</td>
</tr>
<tr>
<td>Gene</td>
<td>386.48</td>
<td>1</td>
<td>386.48</td>
<td>28.11</td>
<td>0.00</td>
</tr>
<tr>
<td>Gender * Gene</td>
<td>93.25</td>
<td>1</td>
<td>93.25</td>
<td>6.78</td>
<td>0.01</td>
</tr>
<tr>
<td>Error</td>
<td>3986.75</td>
<td>290</td>
<td>13.75</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 6.12 shows that, overall, there is a significant difference in anxiety levels within subjects, that is, before and after the gene status information was given (F = 5.38, p = 0.02). Within this, males and females did not differ (F = 0.17, p = 0.68). However, the difference can be attributed to gene status (F = 44.61, p < 0.0001). There was no significant interaction between gene status, gender and receipt of information (F = 0.86, p = 0.36).

Between subjects, there was a significant difference between gene status groups (F = 28.11, p < 0.0001). In addition, the interaction between gender and gene status was significant (F = 6.78, p = 0.01), although there was no significant difference in genders between subjects (F = 0.01, p = 0.91), indicating that the interaction effect was relatively small.

In essence, the most change in anxiety occurs in females. As a group they were more anxious after testing positive for the gene variant than males.

Figure 6.25 shows mean anxiety scores of males and females by gene status and at both time points.
Education

The sample was split into two groups: those with a low level of education (whose highest qualification was A-levels or less) and those with a high level of education (those with a degree or more). A GLM using repeated measures was used to investigate the interactions between level of education and gene status on anxiety levels.

Table 6.13: Differences in level of education in anxiety at baseline and after test results.

<table>
<thead>
<tr>
<th></th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Within-Subjects</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Repeat</td>
<td>41.44</td>
<td>1</td>
<td>41.44</td>
<td>6.66</td>
<td>0.01</td>
</tr>
<tr>
<td>Repeat * education</td>
<td>0.50</td>
<td>1</td>
<td>0.50</td>
<td>0.08</td>
<td>0.78</td>
</tr>
<tr>
<td>Repeat * Gene</td>
<td>348.50</td>
<td>1</td>
<td>348.50</td>
<td>56.04</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gene * Education</td>
<td>2.53</td>
<td>1</td>
<td>2.53</td>
<td>0.41</td>
<td>0.52</td>
</tr>
<tr>
<td>Error (repeat)</td>
<td>1784.80</td>
<td>287</td>
<td>6.22</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Between-Subjects</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Intercept</td>
<td>61882.26</td>
<td>1</td>
<td>61882.26</td>
<td>4412.31</td>
<td>0.00</td>
</tr>
<tr>
<td>Education</td>
<td>7.48</td>
<td>1</td>
<td>7.48</td>
<td>0.53</td>
<td>0.47</td>
</tr>
<tr>
<td>Gene</td>
<td>531.00</td>
<td>1</td>
<td>531.00</td>
<td>37.86</td>
<td>0.00</td>
</tr>
<tr>
<td>Education * Gene</td>
<td>1.49</td>
<td>1</td>
<td>1.49</td>
<td>0.11</td>
<td>0.75</td>
</tr>
<tr>
<td>Error</td>
<td>4025.15</td>
<td>287</td>
<td>14.03</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 6.13 shows that the difference in anxiety levels between baseline and after test results (within-subjects) could not be attributed to level of education ($F = 0.08$, $p =$
0.78). In addition, there was no between-subjects difference in level of education (F = 0.53, p = 0.47).

Ethnicity
A GLM using repeated measures was used to investigate the interactions between white and non-white groups and gene status on anxiety levels.

Table 6.14: Differences in ethnicity in anxiety at baseline and after test results.

<table>
<thead>
<tr>
<th>Within-Subjects</th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Repeat</td>
<td>1.18</td>
<td>1</td>
<td>1.18</td>
<td>0.19</td>
<td>0.66</td>
</tr>
<tr>
<td>Repeat * ethnicity</td>
<td>14.55</td>
<td>1</td>
<td>14.55</td>
<td>2.35</td>
<td>0.17</td>
</tr>
<tr>
<td>Repeat * Gene</td>
<td>137.08</td>
<td>1</td>
<td>137.08</td>
<td>22.15</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gene * ethnicity</td>
<td>0.02</td>
<td>1</td>
<td>0.02</td>
<td>0.003</td>
<td>0.96</td>
</tr>
<tr>
<td>Error (repeat)</td>
<td>1794.54</td>
<td>290</td>
<td>6.19</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Between-Subjects</th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intercept</td>
<td>25890.03</td>
<td>1</td>
<td>25890.03</td>
<td>1840.39</td>
<td>0.00</td>
</tr>
<tr>
<td>ethnicity</td>
<td>0.20</td>
<td>1</td>
<td>0.20</td>
<td>0.01</td>
<td>0.91</td>
</tr>
<tr>
<td>Gene</td>
<td>291.81</td>
<td>1</td>
<td>291.81</td>
<td>20.74</td>
<td>0.00</td>
</tr>
<tr>
<td>Ethnicity * Gene</td>
<td>5.74</td>
<td>1</td>
<td>5.74</td>
<td>0.41</td>
<td>0.52</td>
</tr>
<tr>
<td>Error</td>
<td>4079.64</td>
<td>290</td>
<td>14.07</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 6.14 shows that there was no interaction between ethnicity, gene status and anxiety levels within subjects (F = 0.003, p = 0.96).

Marital status
There were no differences between marital status groups in anxiety levels in either Gene or No Gene groups.

Self-rated health
Because there was only one individual who rated their health as ‘very poor’, this category was eliminated from the analysis. A One-Way ANOVA showed that there were significant differences in baseline anxiety levels between different self-rated health groups (F = 10.77, p<0.0001). Post-hoc Bonferroni analysis (see Table 6.15) indicated that baseline anxiety levels in those who rated their health as ‘poor’ was significantly higher than those who rated their health as ‘average’, ‘good’ or ‘very
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good'. Similarly, those who rated their health as 'average' had significantly higher anxiety levels than those in 'good' and 'very good' health.

Table 6.15: Significant differences in anxiety scores at baseline between self-rated health groups.

<table>
<thead>
<tr>
<th>Mean anxiety scores</th>
<th>Poor</th>
<th>Average</th>
<th>Good</th>
<th>Very good</th>
</tr>
</thead>
<tbody>
<tr>
<td>Poor</td>
<td>17.75</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Average</td>
<td>12.51</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Good</td>
<td>11.22</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Very good</td>
<td>10.20</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* The difference is significant at the p<0.05 level.

Fruit and vegetable intake

A One-Way ANOVA showed that there were significant differences in baseline anxiety levels between different fruit and vegetable intake groups (F = 10.05, p<0.0001). Post-hoc Bonferroni analysis indicated that baseline levels in those who ate less than 2 portions of fruit and vegetables per day were significantly higher than those who ate 4 or at least 5 (see Table 6.16). Similarly, those who consumed 2 or 3 portions per day had significantly higher anxiety levels than those who ate 4 or at least 5 per day. Those differences did not persist after test results had been received.

Table 6.16: Significant differences in anxiety scores at baseline between fruit and vegetable intake groups.

<table>
<thead>
<tr>
<th>Mean anxiety scores</th>
<th>Less than 2</th>
<th>2 or 3</th>
<th>4</th>
<th>At least 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 2</td>
<td>13.76</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2 or 3</td>
<td>12.29</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>10.81</td>
<td>*</td>
<td>*</td>
<td></td>
</tr>
<tr>
<td>At least 5</td>
<td>10.76</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* The difference is significant at the p<0.05 level.

Sugar and fat intake

No differences were found in anxiety levels, either at baseline or after test results, between sugar and fat intake groups.
Activity level

Pearson correlations were carried out to investigate the association between activity level and anxiety scores. Level of activity was very weakly but significantly correlated with baseline anxiety levels (r = -0.16, p = 0.006). The correlation was negative, indicating that higher levels of physical activity tended to be associated with lower levels of anxiety before test results had been received. There was no significant correlation in anxiety levels after test results had been received (r = -0.02, p = 0.68).

Smoking status

A GLM using repeated measures was used to investigate the interactions between smoking status and gene status on anxiety levels.

<table>
<thead>
<tr>
<th>Table 6.17: Differences between smokers and non-smokers in anxiety levels at baseline and after test results.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Within-Subjects</strong></td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>Repeat</td>
</tr>
<tr>
<td>Repeat * Smoking status</td>
</tr>
<tr>
<td>Repeat * Gene</td>
</tr>
<tr>
<td>Repeat * Gene * Smoking status</td>
</tr>
<tr>
<td>Error (repeat)</td>
</tr>
<tr>
<td><strong>Between-Subjects</strong></td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>Intercept</td>
</tr>
<tr>
<td>Smoking status</td>
</tr>
<tr>
<td>Gene</td>
</tr>
<tr>
<td>Smoking status * Gene</td>
</tr>
<tr>
<td>Error</td>
</tr>
</tbody>
</table>

Table 6.17 shows that there was no interaction between smoking status, gene status and anxiety levels within subjects (F = 0.85, p = 0.36). Between subjects, there was a significant difference between smoking status (F = 6.47, p = 0.01) and gene status (F = 18.66, p<0.001). However, the interaction between smoking status and gene status was not significant (F = 1.96, p = 0.16). This indicates that there may be a very small effect of smoking status. Figure 6.26 shows mean anxiety scores of smokers and non-smokers by gene status and at both time points. It appears that smokers tend
to be generally more anxious as a group (ie at baseline) than non-smokers. In addition, smokers told that they did not have the gene variant seem to have higher post-test anxiety levels than non-smokers who were told that they did not have the gene variant.

Figure 6.26: Mean anxiety scores in smokers and non-smokers at baseline and after receiving test results.

6.3.4.2 Perceived risk of heart disease
The following analysis was carried out with the aim of exploring whether perceived risk of heart disease differed according to demographic variables or health indicators.

Age
The 18-20 year old age group was omitted from analyses as it contained only one case. A One-Way Analysis of Variance (ANOVA) with a post-hoc Bonferroni test was conducted on risk perception in age groups. The ANOVA showed a significant difference in baseline perceived risk between age groups (F = 2.32, p = 0.03). However, the post-hoc Bonferroni test failed to specify the location of the difference. This situation occurs because the ANOVA detects differences in variance between the whole sample and each of the sub-samples (in this case, age groups), whereas the Bonferroni test measures differences in variance between sub-samples. There might be a significant difference between one sub-sample and the whole sample, but not
between that sub-sample and any other sub-samples. Nevertheless, the finding is still important.

Gender

A GLM using repeated measures was used to investigate the interactions between gender and gene status on perceived risk of heart disease.

Table 6.18: Gender differences in perceived risk of heart disease at baseline and after test results.

<table>
<thead>
<tr>
<th>Within-Subjects</th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Repeat</td>
<td>3573.71</td>
<td>1</td>
<td>3573.71</td>
<td>16.66</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gender</td>
<td>608.50</td>
<td>1</td>
<td>608.50</td>
<td>2.84</td>
<td>0.09</td>
</tr>
<tr>
<td>Repeat * Gene</td>
<td>12910.57</td>
<td>1</td>
<td>12910.57</td>
<td>60.18</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gene * Gender</td>
<td>179.88</td>
<td>1</td>
<td>179.88</td>
<td>0.84</td>
<td>0.36</td>
</tr>
<tr>
<td>Error (repeat)</td>
<td>67798.09</td>
<td>316</td>
<td>214.55</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Between-Subjects</th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intercept</td>
<td>1108863.00</td>
<td>1</td>
<td>1108863.00</td>
<td>1520.68</td>
<td>0.00</td>
</tr>
<tr>
<td>Gender</td>
<td>1865.36</td>
<td>1</td>
<td>1865.36</td>
<td>2.56</td>
<td>0.11</td>
</tr>
<tr>
<td>Gene</td>
<td>14079.83</td>
<td>1</td>
<td>14079.83</td>
<td>19.31</td>
<td>0.00</td>
</tr>
<tr>
<td>Gender * Gene</td>
<td>164.67</td>
<td>1</td>
<td>164.63</td>
<td>0.23</td>
<td>0.64</td>
</tr>
<tr>
<td>Error</td>
<td>230423.95</td>
<td>316</td>
<td>729.19</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 6.18 shows that, overall, there is a significant difference in perceived risk within subjects, that is, before and after the gene status information was given (F = 16.66, p<0.0001). Within this, males and females did not differ (F = 2.84, p = 0.09). However, the difference can be attributed to gene status (F = 60.18, p<0.0001). There was no significant interaction between gene status, gender and receipt of information (F = 0.84, p = 0.36).

Between subjects, there was a significant difference between gene status (F = 19.31, p<0.0001) but not between genders (F = 2.56, p = 0.11). Further, the interaction between gender and gene status was not significant (F = 0.23, p = 0.64).

Overall, gender does not have an effect on the interaction between gene status and perceived risk of heart disease before or after genetic results are given.

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Figure 6.27 shows mean risk perception scores of males and females by gene status and at both time points.

Figure 6.27: Mean risk perception scores in males and females before and after receiving test results.

From Figure 6.27 it can be seen that females' perceived risk of heart disease is significantly lower than males' at baseline but becomes similar to males' after receiving test results.

Education
The sample was split into two groups: those with a low level of education (whose highest qualification was A-levels or less) and those with a high level of education (those with a degree or more). A GLM using repeated measures was used to investigate the interactions between education and gene status on perceived risk of heart disease.
Table 6.19: Differences in level of education in perceived risk of heart disease at baseline and after test results.

<table>
<thead>
<tr>
<th>Within-Subjects</th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Repeat</td>
<td>3942.73</td>
<td>1</td>
<td>3942.73</td>
<td>18.34</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Education</td>
<td>571.30</td>
<td>1</td>
<td>571.30</td>
<td>2.66</td>
<td>0.10</td>
</tr>
<tr>
<td>Repeat * Gene</td>
<td>13939.22</td>
<td>1</td>
<td>13939.22</td>
<td>64.83</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gene * Education</td>
<td>38.27</td>
<td>1</td>
<td>38.27</td>
<td>0.18</td>
<td>0.67</td>
</tr>
<tr>
<td>Error (repeat)</td>
<td>67302.86</td>
<td>313</td>
<td>215.03</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Between-Subjects</th>
<th>Intercept</th>
<th>Education</th>
<th>Gene</th>
<th>Education * Gene</th>
<th>Error</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1069854.08</td>
<td>532.39</td>
<td>8671.78</td>
<td>2476.21</td>
<td>226482.52</td>
</tr>
<tr>
<td>df</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>313</td>
</tr>
<tr>
<td>Mean Square</td>
<td>1069854.08</td>
<td>532.39</td>
<td>8671.78</td>
<td>2476.21</td>
<td>723.59</td>
</tr>
<tr>
<td>F</td>
<td>1478.54</td>
<td>0.74</td>
<td>11.98</td>
<td>3.42</td>
<td></td>
</tr>
<tr>
<td>Sig.</td>
<td>0.00</td>
<td>0.39</td>
<td>0.00</td>
<td>0.07</td>
<td></td>
</tr>
</tbody>
</table>

Table 6.19 shows that, overall, there is a significant difference in perceived risk within subjects, that is, before and after the gene status information was given (F = 18.34, p<0.0001). Within this, there was no difference between levels of education (F = 2.66, p = 0.10). However, the difference can be attributed to gene status (F = 64.83, p<0.0001). There was no significant interaction between gene status, level of education and receipt of information (F = 0.18, p = 0.67).

Between subjects, there was a significant difference between gene status groups (F = 11.98, p = 0.001) but no significant difference between low and high education groups (F = 0.74, p = 0.39). Further, the interaction between education and gene status was not significant (F = 3.42, p = 0.07).

In essence, educational level does not have an impact on the interaction between gene status and perceived risk of heart disease before or after genetic test results are given.

Figure 6.28 shows mean risk perception scores of high and low education groups by gene status and at both time points.
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Figure 6.28: Mean risk perception scores in low and high educated groups before and after receiving test results.

Ethnicity

A GLM using repeated measures was used to investigate the interactions between ethnic group and gene status on perceived risk of heart disease.

Table 6.20: Differences in ethnic group in perceived risk of heart disease at baseline and after test results.

<table>
<thead>
<tr>
<th>Within-Subjects</th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Repeat</td>
<td>1527.70</td>
<td>1</td>
<td>1527.70</td>
<td>7.07</td>
<td>0.01</td>
</tr>
<tr>
<td>Repeat * ethnicity</td>
<td>102.53</td>
<td>1</td>
<td>102.53</td>
<td>0.47</td>
<td>0.49</td>
</tr>
<tr>
<td>Repeat * Gene</td>
<td>4518.01</td>
<td>1</td>
<td>4518.01</td>
<td>20.91</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gene * ethnicity</td>
<td>273.45</td>
<td>1</td>
<td>273.45</td>
<td>1.27</td>
<td>0.26</td>
</tr>
<tr>
<td>Error (repeat)</td>
<td>68293.19</td>
<td>316</td>
<td>216.12</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Between-Subjects</th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intercept</td>
<td>416344.00</td>
<td>1</td>
<td>416344.00</td>
<td>576.49</td>
<td>0.00</td>
</tr>
<tr>
<td>Ethnicity</td>
<td>4220.80</td>
<td>1</td>
<td>4220.80</td>
<td>5.84</td>
<td>0.02</td>
</tr>
<tr>
<td>Gene</td>
<td>7769.66</td>
<td>1</td>
<td>7769.66</td>
<td>10.76</td>
<td>0.00</td>
</tr>
<tr>
<td>Ethnicity * Gene</td>
<td>167.38</td>
<td>1</td>
<td>167.38</td>
<td>0.23</td>
<td>0.63</td>
</tr>
<tr>
<td>Error</td>
<td>228215.24</td>
<td>316</td>
<td>722.20</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 6.20 shows that there was no interaction between ethnicity, gene status and perceived risk within subjects (F = 1.27, p = 0.26). However, there was a between-subjects difference in ethnic groups in perceived risk of heart disease (F = 5.84, p = 0.02). This indicates that there may be a very small effect of ethnicity. Figure 6.29
shows mean risk perception scores of white and non-white groups by gene status and at both time points. It appears that the between-subjects differences in gene status and ethnic groups as shown by the GLM are that the Gene group have a higher perceived risk after test results than the No Gene group, and that whites tend to have a higher post-test perceived risk than non-whites.

Figure 6.29: Mean risk perception scores in white and non-white ethnic groups before and after receiving test results.

Marital status
There were no differences between marital status groups in risk perception in either Gene or No Gene groups.

Self-rated health
Because there was only one individual who rated their health as ‘very poor’, this category was eliminated from the analysis. There were significant differences between baseline perceived risk scores in different self-rated health groups (F = 12.17, p<0.0001). Post-hoc analysis (see Table 6.21) indicated that baseline perceived risk in those who rated their health as ‘poor’ was significantly higher than those who rated their health as ‘very good’. Similarly, those who rated their health as ‘average’ had significantly higher perceived risk than those in ‘good’ and ‘very good’ health and those who rated their health as ‘good’ had significantly higher perceived risk than those in ‘very good’ health.
Table 6.21: Significant differences in perceived risk at baseline between self-rated health groups.

<table>
<thead>
<tr>
<th>Mean risk perception scores</th>
<th>Poor</th>
<th>Average</th>
<th>Good</th>
<th>Very good</th>
</tr>
</thead>
<tbody>
<tr>
<td>Poor</td>
<td>58.83</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Average</td>
<td>51.49</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Good</td>
<td>42.71</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Very good</td>
<td>26.44</td>
<td>*</td>
<td>*</td>
<td>*</td>
</tr>
</tbody>
</table>

* The difference is significant at the p<0.05 level.

There were significant differences in perceived risk scores after test results in the Gene group between self-rated health groups (F = 5.25, p = 0.002). Post-hoc analysis indicated that perceived risk in those who rated their health as ‘very good’ was significantly lower than those who rated their health as ‘poor’, ‘average’ or ‘good’ (see Table 6.22). However, there were no differences within the No Gene group.

Table 6.22: Significant differences in perceived risk after test results in the Gene group between self-rated health groups.

<table>
<thead>
<tr>
<th>Mean risk perception scores</th>
<th>Poor</th>
<th>Average</th>
<th>Good</th>
<th>Very good</th>
</tr>
</thead>
<tbody>
<tr>
<td>Poor</td>
<td>76.25</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Average</td>
<td>61.39</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Good</td>
<td>61.91</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Very good</td>
<td>45.92</td>
<td>*</td>
<td>*</td>
<td>*</td>
</tr>
</tbody>
</table>

* The difference is significant at the p<0.05 level.

**Fruit and vegetable intake**

There were significant differences in baseline risk perception between different fruit and vegetable intake groups (F = 6.35, p <0.0001). Post-hoc analysis indicated that those who ate at least 5 portions of fruit and vegetables per day had a significantly lower perceived risk of heart disease than those who ate less than 2 or those who ate 2 to 3 portions per day (Table 6.23). However, those differences did not persist after test results had been received.
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Table 6.23: Significant differences in risk perception scores at baseline between fruit and vegetable intake groups.

<table>
<thead>
<tr>
<th>Mean risk perception scores</th>
<th>Less than 2</th>
<th>2 or 3</th>
<th>4</th>
<th>At least 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 2</td>
<td>54.39</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2 or 3</td>
<td>47.00</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>43.04</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>At least 5</td>
<td>35.80</td>
<td>*</td>
<td>*</td>
<td></td>
</tr>
</tbody>
</table>

* The difference is significant at the p<0.05 level.

### Sugar and fat intake

A One-Way ANOVA showed significant differences in baseline risk perception between different sugar and fat intake groups (F = 6.21, p<0.0001). Post-hoc Bonferroni analysis indicated that those who ate sugary or fatty foods every day had significantly higher perceived risk of heart disease than individuals who ate those foods less frequently (see Table 6.24).Similarly, those who ate sugary or fatty foods 3 or 4 times a week had a significantly higher risk perception than those who hardly ever ate those foods. However, these differences did not persist after test results.

Table 6.24: Significant differences in risk perception scores at baseline between sugar and fat intake groups.

<table>
<thead>
<tr>
<th>Mean risk perception scores</th>
<th>Hardly ever</th>
<th>1 or 2 a month</th>
<th>1 or 2 a week</th>
<th>3 or 4 a week</th>
<th>Every day</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hardly ever</td>
<td>24.00</td>
<td>37.47</td>
<td>42.82</td>
<td>44.43</td>
<td>56.77</td>
</tr>
<tr>
<td>1 or 2 a month</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1 or 2 a week</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3 or 4 a week</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Every day</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* The difference is significant at the p<0.05 level.

### Activity level

Pearson correlations were carried out to investigate the association between activity level and risk perception scores. Level of activity was moderately correlated with baseline risk perception scores (r = -0.36, p<0.0001). The correlation was negative, indicating that high levels of physical activity were associated with low perceived risk of heart disease before test results had been received. Activity levels were also
weakly correlated with risk perception after information had been given in the Gene group only ($r = -0.20$, $p = 0.003$).

**Smoking status**
A General Linear Model using repeated measures was used to investigate the interactions between smoking status and gene status on perceived risk of heart disease.

**Table 6.25: Differences in smoking status in perceived risk of heart disease at baseline and after test results.**

<table>
<thead>
<tr>
<th></th>
<th>Type III sum of squares</th>
<th>df</th>
<th>Mean Square</th>
<th>F</th>
<th>Sig.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Within-Subjects</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Repeat</td>
<td>2711.86</td>
<td>1</td>
<td>2711.86</td>
<td>12.71</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Smoking status</td>
<td>50.64</td>
<td>1</td>
<td>50.64</td>
<td>0.24</td>
<td>0.63</td>
</tr>
<tr>
<td>Repeat * Gene</td>
<td>7912.15</td>
<td>1</td>
<td>7912.15</td>
<td>37.08</td>
<td>0.00</td>
</tr>
<tr>
<td>Repeat * Gene * Smoking status</td>
<td>189.95</td>
<td>1</td>
<td>189.95</td>
<td>0.89</td>
<td>0.35</td>
</tr>
<tr>
<td>Error (repeat)</td>
<td>67636.94</td>
<td>317</td>
<td>213.37</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Between-Subjects</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Intercept</td>
<td>791685.41</td>
<td>1</td>
<td>791685.41</td>
<td>1083.86</td>
<td>0.00</td>
</tr>
<tr>
<td>Smoking status</td>
<td>1922.25</td>
<td>1</td>
<td>1922.25</td>
<td>2.63</td>
<td>0.11</td>
</tr>
<tr>
<td>Gene</td>
<td>9506.85</td>
<td>1</td>
<td>9506.85</td>
<td>13.02</td>
<td>0.00</td>
</tr>
<tr>
<td>Smoking status * Gene</td>
<td>46.64</td>
<td>1</td>
<td>46.64</td>
<td>0.06</td>
<td>0.80</td>
</tr>
<tr>
<td>Error</td>
<td>231546.60</td>
<td>317</td>
<td>730.43</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 6.25 shows that, overall, there is a significant difference in perceived risk within subjects, that is, before and after the gene status information was given ($F = 12.71$, $p < 0.0001$). Within this, there was no difference between smokers and non-smokers ($F = 0.24$, $p = 0.63$). However, the difference can be attributed to gene status ($F = 37.08$, $p < 0.0001$). There was no significant interaction between gene status, smoking status and receipt of information ($F = 0.89$, $p = 0.35$).

Between subjects, there was a significant difference between gene status groups ($F = 13.02$, $p < 0.0001$) but no significant difference between smokers and non-smokers ($F = 2.63$, $p = 0.11$). Further, the interaction between gender and gene status was not significant ($F = 0.06$, $p = 0.80$).
In essence, smoking status does not have an impact on the interaction between gene status and perceived risk of heart disease before or after genetic test results are given.

Figure 6.30 shows mean risk perception scores of smokers and non-smokers by gene status and at both time points.

**Figure 6.30: Mean risk perception scores in smokers and non-smokers before and after receiving test results.**

6.3.4.3 Intention to change diet
The following analysis was carried out with the aim of exploring intention to change diet differed according to demographic variables or health indicators.

**Age**
The 18-20 year old age group was omitted from analyses as it contained only one case. Because the scores of intention to change diet are non-normally distributed, it was necessary to use the nonparametric equivalent of the ANOVA, the Kruskall-Wallis test.
No age differences were found in intention to change diet in the Gene group. However, there were significant differences in the No Gene group ($\chi^2 = 15.55$, $p = 0.008$). This test uses the ranks of the data, rather than the actual data and therefore there is no post-hoc analysis. However, Figure 6.31 shows median scores of intention to change diet by age group. 21 to 25 year olds and 36 to 40 year olds seem to be more likely than other age groups to change their diet even though they had been told that they did not have the MTHFR gene variation. As with all nonparametric tests there is an increased risk of a Type II error (not finding an effect which genuinely exists) due to it being less powerful than parametric analyses.

Gender
Differences between genders in intention to change diet was investigated using a Mann-Whitney U test. Differences were approaching significance in the Gene group (median scores in males and females were 75.00 and 84.00 respectively; $U = 4004.0$, $p = 0.07$) but there was no difference between genders in the No Gene group.
(median scores of males and females 62.00 and 56.00 respectively; \( U = 1236.50, p = 0.63 \)).

**Education**
The sample was split into two groups: those with a low level of education (whose highest qualification was A-levels or less) and those with a high level of education (those with a degree or more). Mann-Whitney U analysis showed that less educated individuals were more likely to change their diet than better educated individuals (median scores of 75.00 and 54.00 respectively; \( U = 984.50, p = 0.012 \)), even though they had been told that they did not have the gene variation. In fact, they were almost as likely to intend to change their diet as those who had been told that they did have the gene variation (median scores of low and high education = 82.00 and 80.00 respectively).

**Ethnicity**
No differences were found in intention to change diet between ethnic groups.

**Marital status**
There were no differences between marital status groups in intention to change diet.

**Self-rated health**
Because there was only one individual who rated their health as ‘very poor’, this category was eliminated from the analysis. There was a significant difference between self-rated health groups and intention to change diet in the Gene group (\( \chi^2 = 30.39, p<0.0001 \)). The median score of those who rated their health as very poor was 51.00, poor as 75.00, average as 71.00, good as 84.00 and very good as 97.00. As suggested by Figure 6.32, self-rated health was also found to be significantly correlated with intention to change diet in the Gene group (\( \rho = 0.38, p<0.0001 \)).
Figure 6.32: Median scores of intention to change diet by self-rated health

**Fruit and vegetable intake**

There were no significant differences between fruit and vegetable intake groups in terms of intention to change diet, in either Gene or No Gene groups, or the sample taken as a whole.

**Sugar and fat intake**

Significant differences were also found between sugar and fat intake groups in intention to change diet scores in the Gene group ($\chi^2 = 22.87, p<0.0001$). Median scores for those who hardly ever consumed sugary or fatty foods was 94.00, for those who ate those foods once or twice a month was 90.00, 81.00 for those who ate them once or twice a week, 78.00 for those who ate them 3 or 4 times a week and 63.50 for those who ate those foods every day. The trend indicated in Figure 6.33 that those who consumed less sugar and fat tended to have a higher intention to change their diet was confirmed by a significant negative correlation of $\rho = -0.28$ ($p<0.0001$).
Activity level
Intention to change diet was weakly correlated with activity levels (rho = 0.18, p = 0.007) in the Gene group, but not in the No Gene group.

Smoking status
Differences between smokers and non-smokers in intention to change diet were significant in the No Gene group only (U = 472.00, p = 0.001). Despite being told that they did not have the gene variation which made them at higher risk of heart disease, smokers still intended to change their diet to include more folic-acid containing foods as much as those told they did have the gene variation (see Table 6.26).

Table 6.26: Median scores of intention to change diet

<table>
<thead>
<tr>
<th></th>
<th>Whole sample</th>
<th>Smoker</th>
<th>Non-smokers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene group</td>
<td>81.00</td>
<td>78.50</td>
<td>81.00</td>
</tr>
<tr>
<td>**No Gene group</td>
<td>58.00</td>
<td>76.00</td>
<td>53.50</td>
</tr>
</tbody>
</table>

**significant difference between smokers and non-smokers: p<0.01**
SUMMARY

Individuals with the gene variant showed increased anxiety levels and perceived risk of heart disease, and were more likely to change their diet as a result of the risk information, compared to those without the gene variant.

Individuals who were told they did not have the gene variant showed decreased anxiety levels and perceived risk of heart disease.

Individuals in poorer health, who eat less fruits and vegetables, who are not very physically active or are smokers are generally more anxious.

When told of a gene-based risk of heart disease females or individuals in their early 20s or late 40s tend to become more anxious.

If told they do not have a gene-based risk of heart disease smokers appear to be more anxious than non-smokers.

Individuals in poorer health, who eat less fruits and vegetables, more sugary and fatty foods, or are not very physically active, generally feel at higher risk of heart disease.

When told of a gene-based risk of heart disease white individuals in poorer health appear to feel at higher risk.

When told of a gene-based risk of heart disease those in better health or who eat less sugary and fatty foods, are more physically active or are female tend to be more likely to change their diet as a consequence of the test results.

If told they do not have a gene-based risk of heart disease individuals in their early 20s or late 30s or who are less educated or who smoke are almost as likely as their gene-positive counterparts to change their diet in response to their genetic test results.
6.3.5 To what extent can psychological variables and beliefs predict the impact of genetic health risk information?

6.3.5.1 Anxiety

Multiple regression analysis investigates the relationship between several independent, or predictor variables, and a dependent or outcome variable, based on correlations. Scatter plots were created to check for outliers: none were detected. Any extreme cases may significantly alter the regression model by shifting the gradient of the 'line of best fit', and consequently changing the variables which predict the outcome variable.

The following variables were entered into a Stepwise regression analysis with Anxiety levels after test results entered as the dependent variable:

- Gene status (Gene / No Gene)
- Baseline anxiety levels
- Baseline risk perception
- Risk perception after results

**Psychological variables**
- Belief that diet influences health (before results)
- Belief that lifestyle influences health (before results)
- Belief that genes influence health (before results)
- Belief that diet influences health (after results)
- Belief that lifestyle influences health (after results)
- Belief that genes influence health (after results)
- Monitor score
- Blunter score
- Internal Locus of Control
- Chance Locus of Control
- Powerful others Locus of Control
- Nutrition self-efficacy
- Health value score

**Demographic variables**
- Age
- Gender
- Ethnicity (white / non-white)
- Education

**Health indicators**
- Self-rated health
- Fruit & Vegetable intake
- Sugar & Fat intake
- Physical activity
- Smoker or Non-smoker
The following variables were found to contribute significantly to the regression model (Table 6.27):

**Table 6.27: Variables significantly contributing to the prediction of post-test result anxiety levels**

<table>
<thead>
<tr>
<th></th>
<th>Unstandardised Beta</th>
<th>Standardised Beta</th>
<th>Part correlations (% unique variance explained)</th>
<th>Sig.</th>
<th>R Square</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene or No Gene</td>
<td>-2.91</td>
<td>-0.37</td>
<td>-0.33 (11.2)</td>
<td>0.00</td>
<td>0.25</td>
</tr>
<tr>
<td>Baseline anxiety level</td>
<td>0.38</td>
<td>0.37</td>
<td>0.31 (9.6)</td>
<td>0.00</td>
<td>0.37</td>
</tr>
<tr>
<td>Risk perception after results</td>
<td>0.04</td>
<td>0.25</td>
<td>0.23 (5.3)</td>
<td>0.00</td>
<td>0.43</td>
</tr>
<tr>
<td>Age</td>
<td>-0.27</td>
<td>-0.14</td>
<td>-0.14 (1.9)</td>
<td>0.00</td>
<td>0.44</td>
</tr>
<tr>
<td>Internal Locus of Control</td>
<td>-0.15</td>
<td>-0.12</td>
<td>-0.12 (1.4)</td>
<td>0.01</td>
<td>R = 0.68</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>R² = 0.46</td>
</tr>
</tbody>
</table>

Model:
- Regression: 1655.74
- Residual: 1959.79
- Total: 3615.54
- F: 43.43
- Sig.: 0.00

Having the gene variant or not explains most of the variation in anxiety after receipt of test results (11.2%). Baseline anxiety levels explain 9.6% and risk perception after receiving test results explains 5.3%. Age and internal locus of control explain less than 2% each. All five variables explain 45.8% of the variance of anxiety levels after receiving risk information.

Figure 6.34 shows that the residuals in this model are normally distributed which is essential to verify that the assumptions of regression analysis have been met.
The belief that lifestyle influences health (before test results) was found to be approaching significance in this model \( p = 0.08 \) so this was entered into the regression model with the other five variables, using the Enter method.

However, this lowered the R Square value slightly to .45, and the standardised Beta value was .09 which is low, indicating that the variable was not contributing much to the variance in the dependent variable.

In this model of predicting anxiety levels after receipt of genetic information, age and internal locus of control have been identified as significant predictors. However, they contribute relatively little to the outcome variable: 1.9\% and 1.4\% respectively. Without these variables, the model predicts 42.8\% of the variance of anxiety, compared to 45.8\% of the variance in the model which includes them. This is an increase of only 3\% for the inclusion of two variables.

The Stepwise method of regression can be criticised for including variables in a model simply because they reach a certain mathematical value, as opposed to being included because of theoretical considerations. It is at the researchers discretion to remove variables which contribute little in order to maximise the predictive power of the model with the minimum number of variables. Regression analysis is used here as an exploratory technique rather than an attempt to construct a robust predictive
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model. For this reason it is important to report these variables, however little they contribute, in order to highlight their potential in predicting anxiety levels.

6.3.5.2 Perceived risk

Scatter plots were created to check for outliers: none were detected.

The following variables were entered into a Stepwise regression analysis to predict perceived risk of heart disease after test results:

- Gene status (Gene / No Gene)
- Baseline anxiety levels
- Anxiety levels after results
- Baseline risk perception

Psychological variables
- Belief that diet influences health (before results)
- Belief that lifestyle influences health (before results)
- Belief that genes influence health (before results)
- Belief that diet influences health (after results)
- Belief that lifestyle influences health (after results)
- Belief that genes influence health (after results)
- Monitor score
- Blunter score
- Internal Locus of Control
- Chance Locus of Control
- Powerful others Locus of Control
- Nutrition self-efficacy
- Health value score

Demographic variables
- Age
- Gender
- Ethnicity (white / non-white)
- Education

Health indicators
- Self-rated health
- Fruit & Vegetable intake
- Sugar & Fat intake
- Physical activity
- Smoker or Non-smoker
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The following variables were found to contribute significantly to the model (Table 6.28):

Table 6.28: Variables significantly contributing to the prediction of post-test result perceived risk.

<table>
<thead>
<tr>
<th></th>
<th>Unstandardised Beta</th>
<th>Standardised Beta</th>
<th>Part correlations (% unique variance explained)</th>
<th>Sig.</th>
<th>R Square</th>
</tr>
</thead>
<tbody>
<tr>
<td>Baseline risk perception</td>
<td>0.56</td>
<td>0.55</td>
<td>0.53 (28.2)</td>
<td>0.00</td>
<td>0.29</td>
</tr>
<tr>
<td>Gene or No Gene</td>
<td>-13.26</td>
<td>-0.28</td>
<td>-0.24 (5.6)</td>
<td>0.00</td>
<td>0.47</td>
</tr>
<tr>
<td>Anxiety levels after results</td>
<td>1.88</td>
<td>0.31</td>
<td>0.24 (5.7)</td>
<td>0.00</td>
<td>0.51</td>
</tr>
<tr>
<td>Baseline anxiety levels</td>
<td>-0.97</td>
<td>-0.13</td>
<td>-0.12 (1.4)</td>
<td>0.01</td>
<td>0.53</td>
</tr>
<tr>
<td>Education</td>
<td>2.34</td>
<td>0.10</td>
<td>0.10 (1.0)</td>
<td>0.02</td>
<td>0.54</td>
</tr>
<tr>
<td>Belief that genes influence health (after results)</td>
<td>2.99</td>
<td>0.10</td>
<td>0.10 (1.0)</td>
<td>0.02</td>
<td>0.55</td>
</tr>
<tr>
<td>Sugar &amp; Fat intake</td>
<td>-2.14</td>
<td>-0.09</td>
<td>-0.08 (0.7)</td>
<td>0.05</td>
<td>R² = 0.75</td>
</tr>
</tbody>
</table>

Baseline perceived risk explains most of the variance in risk perception after receipt of results (28.2%). Anxiety levels after the test results, and presence or absence of the gene variation explain over 5% each. Baseline anxiety levels, education, the belief that genes influence health and dietary sugar and fat intake all explain less than 2% each. With all seven variables the model explains 55.6% of the variance of perceived risk of heart disease after receiving genetic test results.

The model as it now stands, however, could be criticised for including variables which contribute very little to the variance in risk perception. Without the latter four variables the model explains 51.3% of the variance in risk perception, compared to 55.6% of the variance when all variables are included. This is only an increase of 4% for the inclusion of four more variables. As mentioned above regression analysis is used here as an exploratory technique rather than an attempt to construct a robust
predictive model. For this reason it is important to report these variables, however little they contribute to the model.

Figure 6.35 shows that the residuals in this model are normally distributed.

**Figure 6.35: Residuals of regression analysis predicting risk perception after test results**
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6.3.5.3  Intention to change diet

Scatter plots were created to check for outliers: none were detected.

The following variables were entered into a Stepwise regression analysis with intention to change diet entered as the dependent variable:

- Gene status (Gene / No Gene)
- Baseline anxiety levels
- Anxiety levels after results
- Baseline risk perception
- Risk perception after results
- Belief that diet influences health
- Belief that lifestyle influences health
- Belief that genes influence health
- Belief that diet influences health after results
- Belief that lifestyle influences health after results
- Belief that genes influence health after results
- Monitor score
- Blunter score
- Internal Locus of Control
- Chance Locus of Control
- Powerful others Locus of Control
- Nutrition self-efficacy
- Health value score

Demographic variables
- Age
- Gender
- Ethnicity (white / non-white)
- Education

Health indicators
- Self-rated health
- Fruit & Vegetable intake
- Sugar & Fat intake
- Physical activity
- Smoker or non-smoker
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The following variables were found to contribute significantly to the model (Table 6.29):

Table 6.29: Variables significantly contributing to the prediction of intention to change diet.

<table>
<thead>
<tr>
<th></th>
<th>Unstandardised Beta</th>
<th>Standardised Beta</th>
<th>Part correlations (% unique variance explained)</th>
<th>Sig.</th>
<th>R Square</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene or No Gene</td>
<td>-9.89</td>
<td>-0.19</td>
<td>-0.17 (2.8)</td>
<td>0.00</td>
<td>0.09</td>
</tr>
<tr>
<td>Nutrition self-efficacy</td>
<td>10.46</td>
<td>0.17</td>
<td>0.15 (2.3)</td>
<td>0.01</td>
<td>0.14</td>
</tr>
<tr>
<td>Risk perception after results</td>
<td>0.33</td>
<td>0.31</td>
<td>0.22 (5.0)</td>
<td>0.00</td>
<td>0.16</td>
</tr>
<tr>
<td>Baseline risk perception</td>
<td>-0.24</td>
<td>-0.23</td>
<td>-0.18 (3.2)</td>
<td>0.00</td>
<td>0.20</td>
</tr>
<tr>
<td>Health value score</td>
<td>-0.94</td>
<td>-0.11</td>
<td>-0.11 (1.1)</td>
<td>0.06</td>
<td>0.22</td>
</tr>
<tr>
<td>Smoker or non-smoker</td>
<td>-9.52</td>
<td>-0.15</td>
<td>-0.15 (2.1)</td>
<td>0.01</td>
<td>0.23</td>
</tr>
<tr>
<td>Belief that diet influences health (before results)</td>
<td>4.56</td>
<td>0.14</td>
<td>0.13 (1.6)</td>
<td>0.02</td>
<td>R = 0.50 R² = 0.25</td>
</tr>
</tbody>
</table>

Model:

| Regression                      | 38159.33            |
| Residuals                       | 114897.20           |
| Total                           | 153056.60           |
| F                               | 12.00               |
| Sig.                            | 0.00                |

Perceived risk of heart disease after test results explains the most variance in intention to change diet (5%), followed by baseline risk perception (3.2%), presence or absence of the gene variation (2.8%), self-efficacy (2.3%) and smoking status (2.1%). Belief that diet influences health and health value explain less than 2% each. With all seven variables the model explained 24.9% of the variance of Intention to change diet.

Figure 6.36 shows that the residuals in this model are normally distributed.
The number of variables in this model, and the little amount of variance they predict in intention to change diet, reflects the complex nature of intentions to perform a behaviour. It could be argued that the model explains little when it contains several variables with such low predictive value. On the other hand, predicting intention to behave is notoriously difficult, and so any variable which predicts intention, however little, is worth knowing about.

When specifically investigating differences between coping style and intention to change diet, a Mann-Whitney analysis showed that high monitors were significantly more likely to intend to change their diet than low monitors (median scores 71.00 and 78.00 respectively, U = 11266.0, p = 0.04). There were no differences between low and high blunters on intention to change diet.

6.3.5.4 Investigating the relationships between genetic information, anxiety, perceived risk and intention to change diet.

A path analysis was conducted to examine the pattern of relationships between genetic information and its impact on anxiety, perceived risk of heart disease and intention to change diet. The three largest predictive variables from each regression analysis described in Section 6.3.4 were entered into the path analysis. Path analysis aims to provide quantitative estimates of the causal connections between variables,
but does not establish causality. The hypothesised relationships make up paths within the path diagram. The negative correlations between gene status and other variables were removed: the correlation coefficients were negative simply because ‘gene positive’ was arbitrarily assigned the lower number for the purposes of analysis.

Standardised (Beta) regression coefficients were used to provide the path coefficients and error (unexplained variance) was calculated by taking the square root of $1 - R^2$ (from the regression model). The total effect of one variable on another was calculated by multiplying the coefficients for each indirect path and summing these together. This figure was added to the coefficient for the direct path between the two variables. Figure 6.37 shows the path analysis with respective direct, indirect and total effects.

Figure 6.37: Path analysis of gene status, anxiety, perceived risk of heart disease and intention to change diet.
Gene status has the biggest effect on intention to change diet. Its total effect is $r = 0.45$. Post-test perceived risk has the next largest impact in intention to change diet ($r = 0.31$). Pre-test perceived risk, gene status and post-test anxiety have similar sized effects on post-test perceived risk. Pre-test anxiety has a slightly smaller effect on post-test anxiety than gene status or post-test perceived risk. Interestingly, there are relatively small effects of post-test anxiety and perceived risk on intention to change diet. It is not possible to establish the causal direction of the relationship between post-test anxiety and post-test perceived risk; it is plausible that either one could cause the other.

SUMMARY

A gene-positive test result, higher baseline anxiety levels, higher perceived risk of heart disease after test results, younger age and a lower internal locus of control were predictive of higher anxiety levels after test results.

A gene-positive test result, higher baseline perceived risk of heart disease, lower anxiety levels before results, higher anxiety levels after test results, higher education, lower dietary sugar and fat intake and a stronger belief that genes influence health were predictive of higher perceived risk of heart disease after receiving test results.

A gene-positive test result, lower perceived risk of heart disease before results, higher perceived risk after test results, higher self-efficacy, lower health value, being a smoker and a stronger belief that diet influences health were predictive of a higher intention to change diet after receiving test results.

Gene status has the largest effect on intention to change diet, followed by post-test perceived risk of heart disease.

Individuals categorised as high monitors, that is, those who actively seek health-related information are more likely to change their behaviour as a result of being given gene-based health risk information.
6.3.6 What influence does information presentation have on the impact of genetic health risk information?

Independent t-tests were carried out to explore whether individuals who received the genetic health risk information which was framed in a positive way (gain framed) differed in anxiety levels, perceived risk of heart disease or intention to change diet from individuals who received the information framed in a negative way (loss framed). However, no differences were found.

**SUMMARY**

There were no differences in anxiety levels, perceived risk of heart disease, or intention to change diet between groups who received gain-framed information or loss-framed information.

6.3.7 To what extent are individuals concerned about the use of genetic health risk information?

Over thirty percent of individuals were not at all concerned about the use of genetic information by other companies or the government (Figure 6.38). About half of individuals were ‘not at all’ or ‘a little’ concerned, and the other half at least ‘moderately’ concerned, with 14% being extremely concerned. The majority of respondents were concerned ‘quite a lot’ about the potential use of genetic information by insurance companies (Figure 6.39). Twenty-five percent were ‘extremely’ concerned. Nearly a quarter were ‘not at all’ or ‘a little’ concerned.
Figure 6.38: Concern about the use of genetic information by other companies or the government.

Concern about the use of information by other companies or government

%: Not at all

0

A little

10

Moderately

20

Quite a lot

30

Extremely

14.3%
Concern about the potential use of genetic information by employers was fairly evenly distributed from ‘not at all’ to ‘extremely’ (Figure 6.40). Thirty-seven percent were ‘not at all’ or ‘a little’ concerned, a quarter of the sample was ‘moderately’ concerned, and thirty-eight percent were ‘quite a lot’ or ‘extremely’ concerned.
Are there demographic differences in the concern about the use of genetic information?

Females were significantly less concerned about the use of genetic information by other companies and the government than males (Figure 6.41: $U = 9924.50$, $p = 0.02$).

Non-White individuals were significantly more concerned about the use of genetic information by employers than White individuals (Figure 6.42: $U = 4487.50$, $p = 0.02$).

Figure 6.41: Gender and ethnicity differences in concern about the use of genetic information by other companies or the government.
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Figure 6.42: Gender and ethnicity differences in concern about the use of genetic information by employers.

Mann-Whitney U tests were performed comparing low and high educated groups on their concern about the use of genetic information by other companies or the government, insurance companies, and employers.

Higher educated individuals were significantly more concerned about the use of information by other companies and the government (Figure 6.43: U = 9691.00, p = 0.01), and about the use of information by insurance companies (Figure 6.44: U = 9529.50, p = 0.01).
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Figures 6.43: Concern about the use of genetic information by other companies and the government in high and low educated sub-samples.

Figures 6.44: Concern about the use of genetic information by insurance companies in high and low educated sub-samples.
There were no significant differences between age groups or marital status in the concern about the use of genetic information.

**SUMMARY**

Individuals tended to be more concerned about the use of genetic information by insurance companies than by employers, and more concerned about the use of information by these groups than by the government.

However, there were demographic differences; females tended to be more concerned about the government than males; non-White individuals were more concerned about employers than Whites; and, high educated people were more concerned about the government and insurance companies than the less educated.
6.3.8 To what extent did individuals understand the genetic and nutritional information given to them?

The majority of individuals understood the genetic information given to them 'quite a lot' (see Figure 6.45). Indeed, over 80% understood it 'quite a lot' or 'extremely' well.

There were no associations between comprehension of information and demographic variables.

Figure 6.45: Comprehension of information

<table>
<thead>
<tr>
<th>Comprehension of information</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>A little</td>
<td>4%</td>
</tr>
<tr>
<td>Moderately</td>
<td>21%</td>
</tr>
<tr>
<td>Quite a lot</td>
<td>41%</td>
</tr>
<tr>
<td>Extremely</td>
<td>14%</td>
</tr>
</tbody>
</table>

SUMMARY

The majority of individuals who took part in the study understood well the information given to them about genes, heart disease, folic acid and their interrelated relationship.
SUMMARY OF QUANTITATIVE RESULTS

Individuals who have been told that they have a gene variant which confers an increased risk of heart disease show increased anxiety levels and perceived risk of heart disease whereas those told that they do not have the gene variant show decreased anxiety levels and risk perception. Moreover, those with the gene show higher intentions to change their diet as a consequence of the test results.

General anxiety and perceived risk of heart disease, in addition to anxiety, risk perception and intention to change diet after receiving test results, can differ in individuals of differing ages, genders and health status.

Demographic factors, psychological variables and health indicators can predict the impact of receiving genetic information about one’s risk of heart disease.

The way health risk information is presented did not appear to affect the impact of the information on individuals.

Individuals were more concerned about the use of genetic information by insurance companies than they were about use by employers or the government, although these concerns differed by gender, ethnicity and education. Individuals generally understood well the information about genes, heart disease and folic acid given to them.
6.4 Discussion

6.4.1 The impact of genetic health risk information

Individuals who were told they had the MTHFR gene showed increased anxiety levels and perceived risk of heart disease, and were more likely to change their diet as a result of the risk information, compared to those without the gene variant. Conversely, individuals without the gene variant showed decreased anxiety levels and perceived risk of heart disease after receiving the health risk information.

Firstly, it is important to note that the increase in anxiety must be taken in context with baseline levels. Both Gene and No Gene groups had baseline anxiety levels below the mid-point of the scale, which indicated that they felt, on average, 'somewhat' anxious. After receipt of information, the Gene group’s anxiety levels were significantly higher but were still below the mid-point of the scale, indicating that they felt between 'somewhat' and 'moderately' anxious. This suggests that the provision of genetic test information relating to heart disease risk does not induce extreme anxiety. The finding adds to the growing body of evidence that genetic information does not necessarily cause distress (eg. Lerman et al 1996; Tibben et al 1997), which is not surprising given its restricted predictive power.

These results confirm that genetically based complex disease risk information prompts a reaction in individuals. Health risk information would be of no use if it did not have any effect. The finding that the Gene group felt at significantly higher risk of heart disease after receiving the test results compared with before, and compared with the No Gene group, corroborates this argument. These individuals felt at slightly lower than average risk of heart disease at baseline (a common phenomenon called ‘comparative optimism’) and after receiving test results reported an above average perceived risk. Assuming that the average individual was at average risk, an ‘above average’ risk (as opposed to a high or very high risk) was a fairly accurate interpretation of the risk information received. However, around 15% of participants rated their perceived risk of heart disease in the highest quartile, that is, upwards of 75 on a scale of 0 to 100. This supports findings from a qualitative study conducted by Bates et al (2003) which reported that the majority of focus group participants
correctly understood the risk implications of "a gene for heart disease", although a minority believed a genetic risk meant that the disease was inevitable.

However, in general, like the anxiety findings, perceived risk was not increased to an extreme extent, suggesting that this type of health risk information has a non-distressing emotional and cognitive impact. This provides evidence against what was suspected by some stakeholders in the qualitative phase of the research.

The greatest potential of genetic tests for complex disease risk factors is to motivate people to take up healthier behaviours, before symptoms present, in order to reduce their risk of that disease. An increase in perceived risk of disease, and, it could be argued, increased anxiety, is necessary for initiating such risk-reducing behaviour change. Perceived vulnerability is a key feature in health behaviour theories such as the Health Belief Model (Janz & Becker 1984) and Sutton (1992) has shown that the amount of fear aroused by a health communication is positively related to the strength of intention to perform the recommended behaviour, which in turn is related to actual behaviour. Previous research has found that distress after genetic testing is related to increased illness-preventative behaviour (Lerman et al 2002).

Intention to change diet to lower the risk of heart disease was higher in individuals with the gene variation than in those without the variation. However, previous research to date has yielded inconclusive findings relating to the behavioural impact of genetic risk information. An overview by Marteau & Lerman (2001) reported some positive impact on the uptake of screening behaviours but not on behaviours associated with reducing the risk of heart disease.

In addition, Senior, Marteau & Weinman (2000) reported that learning of a genetic susceptibility to a disease may actually deter people from engaging in risk-reducing behaviours due to a sense of uncontrollability, or fatalism. However, results presented here did not support this finding. There was no indication that participants who received a ‘positive’ test result showed very high perceived risk of heart disease together with a low intention to change behaviour.
Marteau & Lerman (2001) suggest that behaviour change could be maximised by strengthening two beliefs. First, the belief that the behaviour is effective in reducing risk. In terms of psychological theory this can be likened to Bandura's (1986) concept of outcome expectancy. The study presented here provided detailed information on heart disease, its risk factor homocysteine and the link with folic acid. This information was intended to allow participants to understand the link between genes, diet and disease. It is possible that this information allowed participants to hold a high outcome expectancy of the behaviour.

Secondly, Marteau & Lerman (2001) suggest strengthening the belief in the ability to perform the behaviour. This is most likely referring to the concept of self-efficacy (Bandura 1986). During the experiment, test results were presented with nutritional information which included the recommended daily intake of folic acid tailored to the DNA-based risk, foods which contained high levels of folic acid, and meal and snack suggestions with corresponding percentages of recommended daily intake of folate. It could be that with this detailed information, participants could readily envisage how easy it would be to incorporate folic acid-containing foods into their existing diet, thus reinforcing their sense of self-efficacy.

It is possible therefore that a better understanding of the mechanics behind disease risk, its link with the target behaviour and comprehensive information relating to how to incorporate that behaviour into current lifestyle allows a high outcome expectancy of the behaviour and elevates self-efficacy, leading to the intention to change behaviour.

Some research has reported that test results showing no disease susceptibility genes may result in false reassurance - the perception that one's risk is low if a susceptibility gene variation is not found (Lerman et al, 1996). A number of stakeholders also mentioned this as a potential concern in interviews. Perceived risk after receiving negative test results was found to be significantly lower than perceived risk at baseline in the No Gene group. However, without knowing participants' actual risk it was difficult to tell whether this change reflects false reassurance. The concern relating to this perception is that unhealthy behaviours may be continued if individuals do not see themselves as at risk. However, when asked
how likely they would be to change their diet to help lower the risk of heart disease, the No Gene group rated an average of 57.5 on a scale of 0 to 100, indicating more likely than not. Additionally, there was no indication that participants who received a ‘negative’ test result showed very low perceived risk of heart disease together with a low intention to change behaviour. It could be concluded therefore that this finding echoes that of Marteau et al (2005) who reported that individuals who received a gene-negative result had lower perceived risk, although not to the extent that it could be classed as false reassurance.

In summary, the modest increases in anxiety and perceived risk of heart disease do not support concerns relating to extreme anxiety or fatalism as a consequence of DNA-based complex disease risk information. On the contrary, this level of cognitive and emotional impact is probably necessary in order to motivate risk-reducing behaviour change. Moreover, the impact of the risk information on intention to change diet, even in the No Gene group, challenges concerns of false reassurance and allows the conclusion that this type of genetic-based health risk information could be useful for motivating changes towards healthier a diet without causing negative impact.

6.4.2 Individual differences on the impact of genetic health risk Information

When explored in more detail, differences between individuals were found in how they reacted to the genetic health risk information.

Individuals with self-rated poorer health, those who ate less fruit and vegetables, and those who were not particularly physically active showed higher anxiety and a higher perceived risk of heart disease at baseline. Moreover, these health behaviour variables, together with smoking status, were all weakly to moderately correlated with each other. It is likely that smoking, a lack of physical activity and poor diet contributes to perceived poor health, which in turn produces a perceived risk of illness. Or, that a high trait anxiety leads to smoking and other poor health behaviours. A recent study has confirmed that smoking is associated with illness risk
factors such as inactivity, binge drinking, sleep impairment and poor diet (Strine et al 2005).

Gene-positive females also appeared to be more anxious than gene-positive males after receiving test results. Although this was a small effect it is an interesting one. Females have been found to show higher anxiety in a non-clinical population (Feingold 1994) and social anxiety disorder is more prevalent in women than men (Weinstock 1999). However, there is a lack of research detailing gender differences in the anxiety-related impact of genetic risk information. This may be due the majority of studies focussing on breast cancer and therefore using primarily female samples.

One explanation may be that heart disease is perceived as being more prevalent in males, so being aware of a risk, particularly a risk that appears to be equally likely in both sexes, may come as more of a shock to females, thus increasing their anxiety levels. Indeed, a recent campaign has been launched by the European Society of Cardiology with the aim of raising awareness of the disease in women. Although CVD kills a higher percentage of European women than men and is ten times more common in women than breast cancer it is much less likely to be investigated or diagnosed (European Society of Cardiology 2005).

Gene-negative smokers appeared to be more anxious after receiving test results than gene-negative non-smokers. Again, this was a small effect but interesting to speculate on. The finding is likely to be due to the higher baseline anxiety in smokers, which has been reported previously in the literature (Strine et al 2005). Anxiety is likely to be a cause rather than an effect of smoking, as indicated by a prospective study which revealed that anxiety predicted initiation of smoking in teenagers via an increased susceptibility to peer influences (Patton et al 1998). The possible differences between smokers and non-smokers in terms of the emotional and cognitive impact of health risk information is worthy of further investigation.

There was also a small effect of white individuals appearing to perceive themselves at relatively higher risk of heart disease than non-white individuals when they were told of their gene-related risk of heart disease. Similar results were found by Hughes,
Lerman & Lustbader (1996) who reported that African American women were less likely than white women to report increased perceptions of risk after their relative was diagnosed with breast cancer. However, other findings in the literature are mixed. A study of smokers attending a cancer screening programme found no ethnic differences in cancer risk perception (Ostroff, Hay, Schantz & Maher 2000), and research on the understanding of genetic factors in heart disease reported that African Americans expressed a higher level of perceived risk than European Americans (Bates et al 2003). Ethnic differences in perceived risk of disease and the understanding of risk information is likely to have complex explanations and may be related to culture, socio-economic status and education. There may be implications in terms of the way genetic risk information is presented to different ethnic groups and the level of education or advice that may be required in order to achieve accurate understanding of risk in these individuals. However, without knowing the actual risk of the participants in this study it is not possible to state whether whites or non-whites were more accurate in their perceptions of risk. In addition, consideration must be made for the small proportion of non-white individuals (less than 12%) in this study and the way this may have affected the results presented here.

In terms of intention to change diet, the factors associated with a higher intention in those who received a positive genetic test result were similar to those associated with anxiety and perceived risk. Being more physically active, eating fewer sugary or fatty foods and a better self-rated health were related to a higher intention to change diet. It is possible that these individuals were already health-aware and engaged in healthy behaviours (and consequently felt more healthy), and as a result would be more likely to take on board information and recommendations related to health. Unfortunately, it is individuals who are perhaps less informed about disease risk factors or who currently engage in unhealthy behaviours that would need to be encouraged to eat a healthier diet. Perhaps then, this type of genetic-based health risk information may only be useful for motivating changes towards healthier a diet in already healthy individuals.

Gene-negative individuals who were less educated, and/or who smoked, were almost as likely as their gene-positive counterparts to intend to change their diet. This is encouraging in that the gene-negative information did not discourage this less
advantaged group. It is possible that these sub-groups found the health risk information non-threatening and informative enough to act positively on. The impact of health risk information on different social classes may be an interesting avenue to explore further.

Results also showed associations between psychological variables and the impact of genetic health risk information. Regression analysis revealed that high internal locus of control was a predictor of low anxiety. That is, those who attribute personal responsibility for health-related outcomes were less likely to report anxiety as a result of the genetic information. It is likely that this is due to the sense of control they have over their health. These individuals may be more likely to focus on the controllable aspect of the genetic information (changing the diet to reduce risk) whereas individuals with a low internal locus of control may have focussed on the uncontrollable aspect (the genetic risk factor).

Regression analysis also revealed that a strong belief in the influence of genes on health was predictive of high perceived risk of heart disease after receiving genetic information. Individuals with this belief may have lent more importance to the influence of the MTHFR gene on heart disease than the influence of folic acid in the diet and therefore perceived the disease to be more inevitable than preventable.

Similarly, intention to change diet was predicted by a strong belief in the influence of diet on health. This association is comparable to the link between outcome expectancy and behaviour discussed earlier; the belief that diet can lower the risk of the disease leads to an intention to change that behaviour. One could argue that this belief is in fact requisite for behaviour change. Following on from this, it was discussed before that self-efficacy is also important for behaviour change. Indeed, regression analysis revealed that intention to change diet was predicted by high nutrition-specific self-efficacy.

However, each of these psychological variables predicted only around 2% or less of the variance in anxiety, perceived risk or intention to change diet. Nevertheless, even in this small capacity such variables are important in clarifying the impact of health information and specifically, the explanation of behaviour change. Stronger
predictors were a gene-positive test result and high post-test perceived risk of heart disease. Behaviour is caused by a diversity of biological, psychological, social, emotional, financial and logistical factors and it is therefore important to consider each variable, however minor its role.

6.4.3 Does coping style affect the impact of genetic health risk information?

As discussed in Chapter One, Miller’s theory states that individuals differ in the processing of health information (Miller 1987). Monitors, who actively seek out health information, are more likely to overestimate their perceived risk of a disease and display higher levels of anxiety than blunter, who are more likely to minimise health threats. If these reactions are not too severe this will lead to the uptake of preventative behaviours due to the increased attention to the health threat. However, if the reaction to the information is extreme this may have an adverse effect on the execution of the preventative behaviour.

Analyses showed that there were no differences between low and high monitors or blunter on anxiety levels or risk perception after genetic test information had been received. This fails to replicate findings from several studies showing that (high) monitors experience more distress and anxiety when faced with health threats such as Pap smear examinations and cancer scans than (high) blunter (Miller et al 2001).

One explanation is that the information given was not perceived as a health threat. The monitor-blunter theory holds that monitors experience anxiety when faced with uncertainty and stress. It is possible that the genetic risk information given to participants was not perceived as such, possibly because the detailed nutritional information about reducing the risk of heart disease alleviated any uncertainty or anxiety. Otherwise it is possible that the hypothetical nature of the study failed to produce reactions that would be experienced in real life.

It could be argued that this finding provides evidence against the existence of the monitor-blunter coping style. However, as the theory predicts, analysis revealed that high monitors were more likely to intend to change their diet than low monitors.
Previous research has also shown that monitors are more likely than bluters to engage in risk-reducing behaviours such as cancer screenings (Miller et al 2001).

Although this is encouraging, there is a question of whether this type of information will only encourage monitors, those who already actively seek health information, to engage in risk-reducing behaviour when in fact it should be bluters who are the target population for behaviour change. This is comparable to the point raised in Section 5.4.2 which discussed the finding that individuals who already practice healthy behaviours were more likely to intend to change their diet.

Nevertheless, according to the monitor-blunter theory, these findings indicate that the genetic health risk information provided was sufficiently effective to motivate risk-reducing behaviour, but not too threatening to discourage it.

**6.4.4 What influence does information presentation have on the impact of genetic health risk information?**

Different ways of presenting the health risk information – gain- or loss-framed – did not appear to affect the way individuals perceived their risk of heart disease, felt anxious or intended to change their diet.

**6.4.5 To what extent are individuals concerned about the use of genetic information**

Participants were most concerned about the use of genetic information by insurance companies. Interestingly, during interviews, stakeholders were concerned least by insurance companies. It was felt that genetically-based complex disease risk information was not predictive enough to be used actuarially. However, it was suggested that this issue was covered in the HGC consultation because stakeholders believed that the public were concerned about it. Actually, the focus group research and the internet survey carried out at the time of the public consultation confirmed the public’s concern (People, Science & Policy 2002; YouGov 2003) and findings presented in this thesis also seem to verify it. If the public are genuinely concerned about the misuse of genetic information they may avoid accessing it themselves, and
thus lose out on the potential health benefits. It is important that the public are aware of the genuine risks of data misuse in order to avoid unnecessary concern.

Participants were less worried about the use of genetic information by employers, but the majority still felt moderately or quite concerned. This is similar to previous findings (People, Science & Policy 2002; YouGov 2003). In reality, genetic data for complex disease risk would not be predictive enough to be used by employers either. In fact, it could be argued that if an individual had undertaken such a test, they may be healthier than others as it would indicate an interest and concern about health and a strong possibility that they may be engaging in healthy behaviours, thus actually reducing their risk of illness.

Participants were even less worried about the government or other companies using their genetic data. During interviews, some stakeholders voiced concerns over genetic data being used by the police or by private companies but this was not a widely held view.

After further exploring the data demographic differences in concerns about the misuse of genetic information were found. Males and white individuals were less concerned than females and individuals from other ethnic backgrounds. Slovic (2001) has also reported that men, especially white men, are less concerned about risk. He suggests that this is because they are less willing to express concern, have more knowledge of science and technology and are more trustful of those who run the system.

Individuals with a higher level of education also expressed more concern. For example, 'extreme' concern about the use of information by insurance companies was expressed by around 17% of participants with a low level of education yet by 29% with a high level of education. It could be argued that they have more knowledge of the risks and consequences of the misuse of genetic data, although this conflicts with Slovic's (2001) explanation above.
6.4.6 To what extent did individuals understand the genetic and nutritional information given to them?

The majority of individuals (over 80%) who took part in the study understood well the information given to them about genes, heart disease, folic acid and their interrelationships. This is encouraging given the apparent 'deficit' of public understanding of science (eg. Durant, Evans & Thomas 1992) and the poor opinion stakeholders' held of the public's ability to comprehend risk information. Sturgis et al (2005) believe that although the average citizen may not be knowledgeable of scientific facts he uses everyday experience, values and interests to reach judgments. Participants may not have understood the exact science behind the MTHFR-folic acid-homocysteine-heart disease relationship but they may have understood enough to equate it to other risk factor-illness relationships they did know about and therefore reach adequate understandings and decisions.

On the other hand, the sample used was particularly well-educated which could have biased the results, although there was no association between level of education and understanding of the information.

Although public education on genetic tests for complex disease risks is certainly required, as with any other new technology, it may be patronising to assume the public cannot understand relevant concepts.

6.5 Conclusion

This section summarises the results presented and discussed in this chapter.

- Individuals told of a gene variant related to their risk of heart disease become slightly more anxious and feel more at risk from the disease. This is likely to be related to their increased intention to alter their diet in order to reduce their risk.
- The concern from some stakeholders and evidence from previous research that genetic test information could lead to fatalism or false reassurance was not supported.
Genetic information about complex disease risk factors may therefore be useful in encouraging healthy behaviours without causing a negative impact. However, it may not be effective in individuals who currently have unhealthy habits, or who tend to be disinterested in health issues.

Subtle differences in the way genetic information is presented may have an impact on the encouragement of healthy behaviours in some sub-groups.

Individuals were concerned about the use of genetic information by insurance companies, but less worried about misuse by employers or the government.

The majority of individuals understood well the information given to them about genes, heart disease, folic acid and their interrelationships. This contrasts with stakeholders' poor opinions of the public's ability to comprehend risk information.

6.6 Limitations of the study

The main limitation of this study was that it was a hypothetical situation. The novelty and high cost of genetic tests meant that it was unfeasible to conduct a study using real genetic tests, primarily due to the probable difficulty in recruiting a sufficiently large sample.

The use of the vignette technique was therefore the most practical way of reproducing the situation in which genetic test results are given. However, it does have limitations. It is possible that participants may respond differently to real life situations than those given in the vignette. It is unlikely that many, if any, participants had received genetic test results before and this may have made it difficult for them to imagine the situation. Having said that, the results and corresponding information was well understood by participants and was comparable to other health information relating to a complex disease, such as cholesterol levels. The vignette technique also necessitates participants to make decisions within minutes. In a real situation participants would have been able to ask for advice or clarity from a health professional or other knowledgeable source which may have affected their response to the test results. Until genetic tests are more widely used the
vignette technique is an alternative, and has been used by others when investigating the impact of genetic information (eg Senior, Marteau & Weinman 2000).

Another limitation of the study was that it was measuring intention to change diet, rather than actual change. Using intention as an outcome was obviously a result of a hypothetical research design. Intention to behave is often found to be the strongest predictor of actual behaviour (a recent review cites that it accounts for 28% of the variance: Sheeran 2002), however it is likely that any effect that genetic information had on intention to change diet would be reduced on actual change in diet. A study by Marteau & Lerman (2001) is a case in point. Smokers who were told of a positive genetic test result showing an increased risk for smoking-related lung cancer reported increased intentions to quit yet this did not translate into actual quitting.

Another point to consider is that this study focussed on the intention to include foods in the diet which contained a particular amount of the vitamin folic acid. Adhering to this recommendation may be easier or more difficult, and could involve different factors than, say, reducing the intake of dietary salt or fat, or increasing levels of other vitamins in the diet. This study did not have the capacity to investigate barriers to increasing folic acid intake such as costs and availability of foods, own and other family members' preference of foods and flavours and cultural or religious dietary restrictions.

Notwithstanding different psychological factors, changing eating habits is likely to involve significantly different financial, emotional and logistical factors than those which influence behaviours such as quitting smoking, increasing physical activity or attending medical appointments. For example, quitting smoking involves physiological and complex psychological addiction as well as social factors. Increasing physical activity may depend on the cost of attending exercise classes and available time to dedicate to exercise. Attending health care appointments may depend on distance between home and appointment venue, travel arrangements and time involved in attending the appointment. Therefore, the results presented here may not be generalisable to the uptake or reduction of other health behaviours.
In relation to the sample used in the study, although there was a range of ages, levels of education, ethnic groups and gender, it could be argued that participants were self-selected. In some cases, such as Hewlett Packard and ABPI, individuals were approached randomly and asked to participate in the study. In the cases of Camden Council and Boots plc the research was advertised on the company intranet where employees were invited to participate. Thus, although individuals were approached at random, it was their choice to complete the questionnaire. Those with an interest in health issues may have been more motivated to complete the study. Having said that, if the MBSS scale distinguishes between those who are more attentive to health information than those who are not, the fairly even distributions of high and low monitors and blunters suggests that the sample was not biased in this way.

Moreover, it could be argued that those who were interested in health, genetic testing and/or diet may be those who would be interested in taking a genetic test for complex disease risk factors in reality. At worst therefore, the sample may represent this portion of the population.

Nevertheless, some individuals chose not to complete the questionnaire for some reason, whether it was by not offering themselves as participants or by not returning the questionnaire. It was not possible to obtain any information from non-responders in order to compare them to responders.

In terms of the validated measures used within the questionnaire, one item each was removed from the MHLC-I and MHLC-P scales after testing for reliability and score frequency distribution. This could be criticised as being overcautious on the grounds that it is inappropriate to remove items from validated scales; however, the decision to remove the items was based on the general poor performance of the items indicated by the Cronbach’s alpha values if deleted and the item-total correlations ($r = 0.24$ and $r = 0.11$ for item 8 on the MHLC-I and item 7 on the MHLC-P respectively). Both items were uniformly distributed, and were not felt to be contributing any discrimination to their respective scales; that is, the majority of participants were answering in the same way to the items. The general scale performance is marginally better when these items are removed (Cronbach’s alpha increases from 0.71 to 0.73 for the MHLC-I and from 0.73 to 0.79 for the MHLC-P).
The next chapter will discuss the implications of the findings from the stakeholder analysis and the experimental questionnaire in relation to a wider context in addition to offering conclusions and arguments on the basis of the findings and possible avenues for future research.
CHAPTER SEVEN – CONCLUSIONS
7.1 Suggested future research

7.1.1 Stakeholder analysis

As explained in Section 5.5, these findings provide a snapshot of opinions within this sample, related to the HGC consultation on public access to genetic testing in June and July 2003. It would be interesting to interview stakeholders again in the future to see how, and investigate why, opinions may have changed over time. Or, to interview stakeholders during a similar public consultation on health care technology and compare with results presented here. This may give an indication as to how people react to and cope with new technology and therefore provide information on similar consultations in the future. Similarly, results could be further explored or validated in a larger sample of stakeholders or policy makers using quantitative measures.

Further investigation into the phenomenon of cautious shift amongst a larger sample of public consultation responders or stakeholders would also be valuable. If this phenomenon is true in other public consultations it would have serious implications for the validity of such exercises to provide true opinions.

A semi-structured interview was used to collect data for this study. Future studies may benefit from using a less structured method which may allow for more exploration of explanations, or clarification, of expressed views.

7.1.2 Experimental questionnaire study

The main limitation of this study was that it was a hypothetical situation. Ideally the experiment would have been conducted by providing participants with genetic tests for the MTHFR gene; their test results would have allocated them naturally to the Gene or No Gene group and information framing could have been manipulated for those in the Gene group. Participants would be able to ask for advice or clarity from a health professional or other knowledgeable source which may make their responses different to the hypothetical study presented here.
Another limitation of the study was that it was measuring intention to change diet, rather than actual change. One solution to this would be to conduct a longitudinal study where actual change in diet is recorded. A study like this may also identify other factors, ones which were not measured or considered by the participant when answering the intention to change diet item, which may influence behaviour. For example, the cost of foods which contain folic acid, or how well other members of the family like those foods.

This study investigated the impact of dietary recommendations given on the basis of a genetic test result. Future studies could compare genetic information with non-genetic information such as cholesterol level or a family risk indicator with intended dietary change as the outcome measure. One of the criticisms stakeholders gave for genetic tests for complex disease risk factors was that they give no more information than other medical tests. It would be worth comparing the impact of gene based and non-gene based risk information in order to investigate this argument.

Another point to consider is that this study focussed on the intention to include foods in the diet which contained a particular amount of the vitamin folic acid. Further research could be conducted where the behavioural outcome measure is, for example, reducing the intake of dietary salt or fat, or increasing levels of other vitamins in the diet. Results could then be compared with those of the present study. Moreover, factors which are likely to affect whether nutritional advice is taken on board such as costs and availability of foods, own and other family members’ preference of foods and flavours and cultural or religious dietary restrictions could also be investigated, possibly through qualitative analysis.

Research on the potential impact of genetic information on other health-related behaviours is likely to be required in the future as gene-disease associations become more precise and predictable. For example, a study investigating the impact of personalised medicine on regimen adherence would be helpful considering the forthcoming introduction of pharmacogenetics.
7.2 Implications for public health and regulatory policy

In order to conclude the thesis and discuss implications for public health and regulatory policy three key findings from this research are highlighted:

- Complex disease risk information can encourage healthy behaviour intention, albeit to a greater extent in those already motivated to be healthy. No negative impact from this information was apparent in this research, despite concerns raised by Senior, Marteau & Weinman (2000).

- Contrary to some groups’ concerns, stakeholders were not worried that genetic information about complex disease risks would have any negative consequences in terms of employment or insurance discrimination. Thus access to genetic information for complex disease risk does not raise issues which are not already apparent from, say, knowing information about one’s Body Mass Index or cholesterol level.

- Stakeholders were concerned that the public cannot comprehend genetic and/or risk information, but results presented here show that participants understood well the information given to them. In addition, stakeholders showed a ‘cautious shift’ in decision making. This could be related to a reluctance to hand over control of health to the individual or, alternatively, it may reflect a genuine concern to protect a vulnerable minority of the population from potentially complex information.

To address the public health concerns of today individuals’ autonomy over their own health should be respected so that they can actively choose healthy lifestyles. This is accepted and encouraged by health policy makers, illustrated clearly by recent Department of Health programmes and documents such as The Expert Patient, The NHS Improvement Plan and the Choosing Health white paper. It also seems to be a vision accepted by the public. British citizens are increasingly becoming active partners in decisions about their medical care. The Internet has enabled them to be
more informed about health matters and they are increasingly being consumers of health care rather than patients.

In order to achieve this autonomy, access to, and education about, health information to aid health-related decisions is vital. Genetic testing for risks to complex diseases is one such source of information. Genetic testing is currently only available via a medical professional or by private companies. The recent public consultation led by the HGC investigated other options of access to genetic services. Stakeholders interviewed for this thesis expressed concern that the public are not capable of understanding the predictive power of genetic information, or indeed concepts of risk in general, and it is this perception that may have led to the HGC recommendation that genetic tests should not be freely available.

It may also be due to a reluctance of some health professionals to hand over health decision-making power to the individual. This unwillingness to allow citizens autonomy over their health may be related to the recent (late 20th and early 21st century) decline of the medical profession, as discussed by McKinlay & Marceau (2002). They attribute this to several factors. One is the emergence of other health professionals such as nurses, complementary providers and those from specialist disciplines such as optometrists and anaesthetists. These professionals can now provide medical care which was once only in the domain of the doctor. Extended and supplementary prescribing by nurses and pharmacists is one example of this in the UK.

A second factor is the 'information revolution'. Public access to health information via the Internet has increased levels of public medical knowledge, and while empowering patients, has reduced the wisdom-related status of doctors, which in turn has changed the doctor-patient relationship and enforced the provider-consumer model. Moreover, access to computerised medical records and publicly available data on the performance of medical facilities and practitioners has allowed doctors to come under scrutiny from patients.

However, it may be that stakeholders are genuinely committed to public welfare and are reluctant to allow this information to be freely available without the provision of
appropriate support. This argument is supported by the initiative to establish a Centre
for Genetic Education to support the education and training of health professionals in
the field of genetics in order to provide the public with a reliable and unbiased
source of accurate information and support (Burton 2003).

Research presented in this thesis has indicated that individuals given this information
about their health are motivated to alter their diet in order to reduce their risk of
disease with no negative impact. That is, the moderate increase in anxiety seen in
participants was sufficient to encourage behaviour change intention, but was not
extreme. Results also show that participants who received a ‘positive’ test result did
not show very high perceived risk of disease together with low intention to change
diet, nor did ‘negative’ result individuals show very low perceived risk of disease
together with low behaviour change intention. Previous findings of fatalism and false
reassurance respectively are therefore not supported here (Senior, Marteau &
Weinman, 2000; Lerman et al 1996) but are similar to those found for example by
Marteau et al (2005) who found that the changes in perceived risk were not sufficient
to be classed as false reassurance. The case for genetic exceptionalism therefore does
not seem appropriate in the context of complex disease risk information.

There appears to be no real concern that this information could cause harm in terms
of employment or insurance discrimination. It could be argued that strict regulation
over access to this information would be a paternalistic decision; a result of a lack of
trust in the public to understand and use genetic information. Such an outcome could
in turn serve to generate or perpetuate public misperceptions about the role of
genetics in complex disease causation. In addition to this, or linked to this, it could
also be argued that strict access regulation is an indication of the medical profession
attempting to retain their ‘special’ status in society.

The finding that stakeholders tended to show a ‘cautious shift’ in decision making
could be related to this. This indicates that although public consultations, including
that undertaken by the HGC, may on occasions produce valuable insights, there is a
danger that the methods they employ will generate systematically biased findings.
Individuals expressing their own opinions were less inclined towards access-related
regulatory restrictions than they were when responding as representatives of
organisations. Policy makers should therefore be aware of the limitations of such consultations. They should be prepared to invest in research to validate and explain the opinions and assumptions of individuals asked to act in expert or representative roles.

The main consequence of over-regulation is that the public would not have easy and convenient access to information which may be beneficial in motivating health risk-reducing behaviours. The argument that there is currently weak evidence to link most gene variations to diseases is unconvincing when used against free access. Many products or alternative therapies such as homeopathy or reflexology are unregulated and their health benefits or consequences are little known. Yet they are freely available to the public. The public should be made aware of the predictive limitations of complex disease risk genetic tests, through mass public health education, together with resources to interpret, understand and use the information properly.

Over-regulation may also suppress health technology innovation and development which could force the industry to move out of the UK. A cautious approach is not conducive to embracing scientific breakthroughs and technological advances.

However, the government response to the HGC recommendations is still forthcoming at the time of writing. It is possible that the government does not agree with the HGC recommendations and is allowing time for genetics to seep into the public consciousness before regulatory decisions are made.

There are possible solutions to the tension between allowing free access to information and protecting those who may be vulnerable to any harm it may cause. This is termed ‘asymmetric paternalism’. A regulation may be acceptably paternalistic if it protects a minority of citizens who are at risk from making errors, while imposing little or no harm or inconvenience on the majority of rational citizens (Camerer et al 2003). In real life terms this could be allowing genetic tests for complex diseases to be accessed via community pharmacies, similar to pregnancy testing, in which a health professional with adequate training and knowledge would be available, if wished, to provide interpretation and advice on the results of the test.
and on subsequent risk reducing actions. This approach would protect a small minority of the population who may be at risk of misinterpreting genetic information and experiencing harm as a consequence. A similar concept of ‘pragmatic paternalism’ was introduced by Duggan (1998) who found that some patients desire information about their medicines and some would prefer not to know too much. A pragmatic paternalistic approach - removing barriers between patient and health professional whilst ensuring maximum patient benefit – would cater for patients who want autonomy at the same time as accommodating those who do not.

This would of course require pharmacists to undergo training and education in aspects of genetics and genetic testing. Recommendations for this have already been put forward by Burton (2003) which included the need for teaching in basic science, clinical aspects and ethical, legal and social aspects of genetics as well as providing accessible learning materials for professionals already in practice. The establishment of a Centre for Genetics Education and a formal programme for genetics education was also suggested. Having access to a trained health professional would allow genetic test results and its consequences to be accurately interpreted and explained and would allow recipient’s misconceptions to be corrected, thus addressing concerns held by stakeholders about the public’s lack of understanding of concepts of risk.

In conclusion, policy makers, health professionals and the public need to recognize and embrace the shift in health care and plan for a future where the individual has autonomy over health decisions which he or she is capable of making, and where information to aid those decisions is easily accessible. Health professionals have a duty to help interpret this information and provide support whilst defending the freedom and well-being of individuals. Without this balance public health could become a controlling rather than an enabling discipline.
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APPENDICES
## Appendices

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Thank you very much for taking the time to see me.

The interview should take no more than half an hour.

1. Can I just start by asking a question about you? If you had to say what your primary profession or occupation was, how would you describe yourself?

2. If the subject of public access to genetic testing in general is raised, what are the most important issues which immediately come to mind?

3. The research I am doing is about access to (and regulating) testing for vulnerability to complex diseases. That is, testing for multiple rather than single gene indicators of risk of contracting conditions such as heart disease, a cancer or forms of mental illness. What do you see as the key issues in this area?

4. I'm going to show you a checklist of some issues and concerns. In relation to deciding on regulatory policy on public access, could you please indicate the importance of these?

5. The Human Genetics Commission recently held a stakeholder consultation on genetic testing. Were you aware of, or did you or [your organisation] contribute to this consultation?

6. Do you have any comments on this consultation?

7. What are your opinions on the recommendations made by the HGC?

8. In order to keep this interview to time, I am now going to show you two pairs of statements relating to modern health care in general and the development of genetics in general. Please indicate your opinion, as a representative of [your organisation].

9. Now I would like you stand back from your role as (job title). Speaking just for your self as an individual with your own private personal beliefs about what is best for you and your family, could you look at the statements again, and indicate your position as a private citizen?

10. Thank you. Returning now to you in your professional role, could I ask whether you believe that genetic information about health risks is different from other types of potentially predictive information about health, like family history or weight or smoking?

11. Thank you. For the final part of the interview I am going to give you a list of statements, and ask you to indicate the extent to which you agree or disagree with them.
12. In respect of any of the statements in this final section, were there any in which you felt a particular tension between you as individual and you as a representative of (name stakeholder group?)

13. Thank you. Is there anything else you would like to add or comment?

14. Is there anyone you can recommend that would be useful for me to interview?

## QUESTION 4

Please indicate the importance of these issues in relation to deciding on regulatory policy on public access.

<table>
<thead>
<tr>
<th>Concern/Issue</th>
<th>High importance</th>
<th>Moderate importance</th>
<th>Low importance</th>
</tr>
</thead>
<tbody>
<tr>
<td>Test information could be used to determine or trace paternity</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Test information might not be accurate, and/or statements about error rates might not be understood</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Test information could be used to determine or trace individual identity</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Test information might cause anxiety</td>
<td></td>
<td></td>
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<tr>
<td>Test results may lead to discrimination and/or a 'genetic underclass' if disclosed to insurance companies or employers</td>
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<tr>
<td>Test information might evoke false reassurance about health</td>
<td></td>
<td></td>
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<tr>
<td>Tests could be done on children or others unable to give informed consent</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Test information might cause people to become fatalistic about their health</td>
<td></td>
<td></td>
<td></td>
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</tbody>
</table>
QUESTION 8

As a representative of (named organisation/stakeholder group), where do you stand on these spectra?

Individuals have a right to free access to information about their genetically determined health risks. All genetic tests which are physically safe to use should be available for people to buy freely as well as being available through the NHS

Adults are responsible for their own health. We need to move on from professional paternalism to more emphasis on individual choice and autonomy in all areas of health

The public needs protection from exploitation and needless anxiety. Tests should only be available through the medical profession

People at risk of illness are often the most vulnerable in society. They are in need of informed professional direction backed by comprehensive government regulation
As an individual, where do you stand on these spectra?

Individuals have a right to free access to information about their genetically determined health risks. All genetic tests which are physically safe to use should be available for people to buy freely as well as being available through the NHS.

Adults are responsible for their own health. We need to move on from professional paternalism to more emphasis on individual choice and autonomy in all areas of health.

The public needs protection from exploitation and needless anxiety. Tests should only be available through the medical profession.

People at risk of illness are often the most vulnerable in society. They are in need of informed professional direction backed by comprehensive government regulation.
## QUESTION 11

Please indicate the extent to which you agree or disagree with the following statements.

<table>
<thead>
<tr>
<th>STATEMENT</th>
<th>Strongly agree</th>
<th>Agree</th>
<th>Neutral</th>
<th>Disagree</th>
<th>Strongly Disagree</th>
</tr>
</thead>
<tbody>
<tr>
<td>1  If the public could freely buy genetic tests for health risks this would lead to unnecessary demands on NHS resources</td>
<td></td>
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<td>2  Expert counseling before and after testing for vulnerability to complex genetic conditions is essential</td>
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<td>3  If restrictive approaches to controlling the sale of genetics based tests to the public drives entrepreneurs out of Britain, this country is likely to lose more than it gains</td>
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<td>4  If providing genetically based information about complex disease risks were to reinforce awareness the dangers of factors such as obesity, smoking or poor diet, this would be a good reason to allow the free sale of relevant tests</td>
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<td>5  Most people will never be able to understand the nature of genetically mediated health risks for common conditions – they will assume that if you have ‘the gene’ you will inevitably get the disease</td>
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<td>6  Advertising prescription only medicines is inherently undesirable</td>
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<td>7  The fact that people can order genetic tests on the internet and send them off to the US for analysis makes it impractical for this country to try to impose more stringent regulations than the Americans</td>
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<td>8  Pharmacies rather doctors surgeries would be the best place to provide genetic tests for complex disease risks</td>
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<td>9  Groups such as the elderly are particularly vulnerable to exploitation by commercial organisations selling tests for disease risks</td>
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<td>10 A stringent regulatory approach based on scientific evidence is vital – if the links between particular genotypes and the risk of an illness are only theoretical companies should not be allowed to sell tests to the public</td>
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<td></td>
<td>There is too much emphasis on regulation and too little on enabling the public to use new sorts of information effectively</td>
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<td>11</td>
<td>In a free society people have a right to normal access to health information and diagnostic and other tests which are physically safe to use</td>
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### Annotated interview transcript

<table>
<thead>
<tr>
<th>SC</th>
<th>When you think about genetic testing being available to the public, what are the immediate issues that spring to mind for you?</th>
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<tbody>
<tr>
<td>P</td>
<td>Available to the public via the NHS?</td>
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SC Well, broadly... the discussion that the HGC were having was *how* available should they be – should they be more available over the counter, or via a pharmacists or what sorts of tests should only be available through the NHS, so what did you think the main issues were?

P Well I think there’s different categories of genetics tests. There are tests in the area of cancer where we know that a single gene defect can very much significantly alter the risk of inheriting certain cancers, or developing that cancer. But they are very few. As far as we know there are breast and ovarian cancer and bowel and a few related cancers, and there might be something going on with prostate and pancreas but we don’t know the exact gene mutations yet. Now these tests tell you an awful lot about yourself – if you test positive for that test you learn something rather upsetting about yourself – that’s one category of test. And then there are categories of tests which I think are qualitatively quite different because they don’t tell you very much about yourself. The tests that you can buy at Boots or the one they were trying to sell through Body Shop, they tell you something about things they call your metabolic profile or whatever, the information you gain from these tests is very limited, to use refrained language. I’d say they are actually a case for the advertising standards agency because they make claims that they can’t actually fulfil. They proclaim they tell you something about your cancer risk which they just blatantly don’t. But these tests might obviously be blurred - there are some tests which test for some protein to do with prostate and we don’t quite know whether it mean your increased risk or whether you’re on the way there, so these categories are still blurred but its still quite a useful way to think about it. And the ones that test for single gene mutations that very significantly increase your risk, they obviously, I think its quite obvious, that they need to be delivered in the context of good reliable information and also a space being provided for the patient where the patient can think through the implications of going for the test. Some commercial companies might claim that they do that, that they provide that space. I’m quite critical of pharma companies telling us they’re in the business of information provision. Of course they also provide information, everyone’s in... |

| Impact of knowledge of genetics |
| Validity and usefulness of complex disease risk tests |
| Information, support & counselling |
| Perception of industry sector |
the business of providing information, but what kind of information? And how useful is that information to people who are quite desperate and anxious and often for no good reason and often good impartial information will be ‘don’t worry and don’t waste your money on a genetic test’. That’s not the information that they give because they want to sell a test. So I think in order to provide that space in order for people to really reflect upon the implications of the test and also to give them the information they need to read the test result properly in the contexts of their own lives, the probably need to have something that resembles genetic counselling. Now, you might not need to call it that, and I think the term ‘counselling’ is a little bit dubious in that context anyway, but we need to basically...the patient needs to have the opportunity to sit down in a room with someone who doesn’t have an interest in pushing the test or not pushing the test, and reflect upon what this means to their own lives and their families. And the questions that need to be reflected upon range from ‘how would I feel with this or that test result’ because quite a number of people want to get tests to learn they’re OK and it needs to be...it sounds a bit patronising, but it probably needs to be explained that you might also learn you’re not OK and it’s a very convenient way of conceptualising genetic testing that people might use when they’re very stressed about cancer running in their families – ‘I’ll get the test and then I’ll learn that I’m OK’ but they might do the test and learn they’re not OK. So that’s one thing that they need to think about – what would you do if you learned you had a breast cancer risk of 80%, have you thought about the implications of that, are there some options available that you’d certainly go for or not go for – those kinds of thoughts. Then there is all the familial consequences – who in your family would you have to tell, who would it also affect, how would you cope with that, at what point would you want to tell them. Then we also always tell people that they might learn something about their family that they didn’t set out to find out so people find out that they came from adoption, and its just worth having a fleeting thought about that before you embark on this. I might find out my dad is not my dad – it might be worth dropping that into your family conversation – are we all sure about how related we actually are before we...that kind of thing. Then there’s insurance implications – we’ve got a moratorium that runs out in 2006 and we always say to people no-one knows what’s going on after that. Most people who now go for genetic testing will have a long life span after 2006 so they need to think about that. And there are other issues of discrimination and data protection that if we’re honest with ourselves we can’t give people good information on because its all very unclear. So now in the new genetics White Paper they’re saying they want to do something about genetic discrimination but there will be exceptions for the police and already research databases are fully protected so if you give your information, your genetic material into a research programme, which almost everybody does who...
gets a genetic test, this data is not entirely protected. Now, I don't think we need to be desperately worried about that because researchers so far have got a very good track record of being safe with information – I can't think of any leak that has occurred in the context of NHS clinical research – and that’s to do with the fact that these people can be trusted I think. I don’t know whether the Home Office can be trusted and people feel different about the Home Office. So I don’t think we’re trying to worry people too much about these issues but its just something that they need to know that the police and the courts and the Home Office will have access to that data. So these are the issues we need to flag up to people I think when they’re contemplating a genetic test and maybe most importantly people need to know that the vast majority of tests return inconclusive results. I met some genetic specialists yesterday and they said, when you press them, and they’re all very enthusiastic about things, only between 10 and 20 percent of families actually get a positive test result and that is in the context of high risk families. So people need to know when they embark on all of this that the most likely outcome is that they won’t learn anything useful. So people, having told they’re high risk because of their family background, and having decided they want to go for genetic testing in order to be able to drop out of screening which say, for bowel cancer is very unpleasant, that’s not happening. If you don’t have the cancer yet you go for mutation searching, you either get a positive test result which means you’ve got the gene mutation, or you get an inconclusive test result which means we don’t know whether you’ve got a mutation but because you’ve got that family history means we advise you to continue with the screening. So in either case you have to continue with the screening. So just when you explain all of that to people, people suddenly notice that a genetic test isn’t the most important step of the whole process. The most important step is to look at family history and give them advice on whether they should get more regular screening than other people.

SC So those sorts of issues, are they just for the BRCA type gene mutation tests or do you think they apply for the ones you mentioned earlier – the nutrition metabolism type tests as well?

P I honestly think the majority of these kinds of tests for God knows what kind of profiles, we going to look at in a hundred years time, just as we look at other things from the Victorian era – use this gel and grow curls, you know, its just one of those things, it’s the early stages of the signs developing and people make these claims about them, So I think the vast majority of those will just land on the rubbish heap and we’ll laugh about that. But there might be some serious stuff in there – there might be some serious stuff about prostate cancer that we could find out. And also people might go for a test that at the moment doesn’t tell us anything but maybe in 20 years time we’ll know that people who have tested
positive for this or that have a higher risk of Alzheimer’s or higher risk of Parkinson’s or a higher risk of depression – things that people really did not set out to find out about. So because this is an evolving science the fact that we now kind of think these things are so useless, they’re harmless, doesn’t mean they actually are harmless. So someone might hold information about you afterwards and that’s the other thing that we don’t know about these companies – what are they doing with the information they get, that in 20 years time might actually tell you quite a lot about yourself. At the moment it doesn’t so at the moment its just a waste of money but in 20 years time you might learn something upsetting about you. So I think they do still require regulation, these kinds of tests, but I don’t know whether we need to prohibit access to them so I think we might have to get the Advertising Standards Agency and say ‘what are the claims you making? Can you substantiate them? If you can’t you have to take this off the shelf or make a different claim about the product’.

SC So it’s the evidence base then is the main issue for those types of test?

P Yeah, I think it is.

[SC explains question 4]

SC Talking about the HGC consultation, was [your organisation] invited to respond to that?

P We were, actually I don’t know whether we were officially invited, but I only started working here when this was all way under way so we didn’t actually respond because no-one else was actually covering the genetic issues for us.

SC Were you aware of who they did ask and the process – have you got an idea of whether you felt it was fair and unbiased.

P I don’t know.

SC Do you know about the recommendations that they made – that sort of tiered approach? What do you think of that? Do you think that’s appropriate?

P What do you mean by the tiered approach?

SC They came up with something similar to how medicines are regulated and they put serious, single-gene Huntington’s type tests in the high category where it would only be available through the NHS or specialists with advice and then at the other end it was over-the-counter scenario for these life-style, nutrition type tests, then other things in the middle. So quite a flexible graduated
approach. Do you think that was appropriate?

P There is some truth in that but as I said already that the things that we now think don’t tell us very much might in the future tell us more. Also I think there is a need for regulation even with those that we think don’t tell us very much because we need to know what these people do with the data they’ve got. Because you can trace individuals back to their sample, that’s different from a urine sample or pregnancy or... they’re comparing them with those kinds of tests and I think there’s a good case not to single out genetic information and say ‘ooh it’s so scary’ and we need to get in there with the heavy-handed regulation necessarily but we need to think where is it different and I think if you extract DNA you can trace that back to the person, you can test it for anything else that you come up with in the future and I think there is a need for regulation for companies who provide those tests need to show what they do with those samples and probably they should destroy them after they’ve done the test, which they’re not going to do because they’re interested in getting these databases together. So I think there is a need for regulation on all these levels but I think it is useful to think across all the range of genetic testing that there is. And I think you can read the report two ways - on one hand it flags out all the concerns and it says there are some very serious tests and they need more serious regulation, but it doesn’t actually provide a very firm recommendations on what should happen – it just says it needs to be contemplated and we advise that the government thinks about it, and its just... it kind of gathered the information and brought it together and it set it out like this but then it fizzles out into this ‘wouldn’t it be nice if we thought about this’. And I think there is a problem with the HGC and their role and that they don’t have much power but they could have come out with more committed recommendations and they could have said ‘we think it takes primary legislation for those, and we think it takes the Medicines Control Agency to look at these and it takes the Advertising Standards Agency to look at these and we expect all these bodies within the next 12 months to come up with regulation’. I think its completely non-committal and I think that’s a wasted opportunity.

SC Just going back to what you said earlier and what you mentioned just now about genetic information being different to any other information. How do you feel about...that was an argument that genetic information is much more important or predictive or scary than any other – family history, smoking or diet. How do you feel about genetic information?

P I don’t think we need to make out it is such a special case. For example, in the area of cancer, people say the difference with this information is that it tells you something about your relatives and there’s obviously a truth in that, but it’s also true that normal
cancer doesn’t have any implications for the wider family either. It’s like that with health information anyway, if effects the whole family - people have to come to terms with it as a family often, whether it’s an inherited disease or not. So there are some artificial distinctions being made up I think and there is a case for saying yes genetic information has some distinct qualities but if your wife or your daughter or your mum has just been diagnosed with cancer, this has implications for you as well, it might have genetic implications but it also has other implications. But I think there are... because it is an evolving science we need to be careful to protect in a way the consent people have given because people consent to a certain test being run and certain information being given but because this is an evolving science this sample could be used for so much different stuff and I think we need to be careful of that. At the moment if you give your blood sample to be tested for BRCA1 or 2 as far as I know, all the hospitals say ‘if we find more genes we’ll run your blood sample through these tests as well. So for BRCA3 we’ll run it through that as well. People consent to that coz that makes sense – they want to find out if they carry a breast cancer gene but what they don’t consent to is if BRCA3 maybe only increase your breast cancer slightly but also increases your risk of schizophrenia quite substantially – that is hard to explain to people. And you just need to think about what have people actually consented to. So I don’t think we just need to march in there, clamp it all down and say ‘this is really dangerous knowledge and we mustn’t treat it like any other knowledge’ but I think we need to kind of keep on our toes as to how genetic knowledge does differ on occasions and people feel differently about for example, the implications for insurance and employers. Because that’s one thing in find people are very worried about – the implication for insurance. Then again, people who have got that kind of a family history that qualify for a genetic test – BRCA1 or 2 or bowel cancer – they won’t get health insurance anyway just because of their family history. So its just another way that health insurers have asked, since the beginning of time, about your risk. And they ask you did your parents die of heart disease before they were 60, do you have other relatives who had heart disease, and its just another way of getting the sample information. So people are worried about it and I think we need to acknowledge that and think about how we need to protect them but its not that different from what is going on anyway.

SC Is that the same do you think in terms of diagnosis for GPs, that the genetic information, will that give any more information than they will have got from family history or lifestyle behaviour?

P I think we will learn a bit more, well quite a lot more I suppose, about a small number of families where something clearly is going on and that can be useful for primary care and then the vast majority of our knowledge of genetics will be about things
that on their own will increase your risk ever so slightly and if they come into combination with another 25 genes might actually push you beyond the threshold where you’re more likely to get stomach cancer or whatever. But I think that’s beyond what primary care can deliver – that kind of knowledge model. People will need the brain the size of a planet to be able to do that, and even more so if you then try to correlate that with lifestyle. So I think we’re just deluding ourselves that GPs can accurately, will be able to accurately tell you whether the combination of these 300 genes and your exercise and your smoking will push you beyond this barrier. At least I don’t think that’s likely to happen.

[SC explains and participant completes questions 8, 9 and 11]

[END]
The University of London School of Pharmacy is investigating attitudes towards health, and how information about our genes and lifestyles can protect us from illness.

By filling in a simple questionnaire you will be contributing to important research to help the fight against cancer, heart disease and other illnesses.

Everyone replying will be automatically entered into a prize draw to win £100!

The questionnaire can be emailed OR posted to you - whichever you prefer. It will take about 20 minutes to complete.

Email sarah.carter@ulsop.ac.uk to be part of this valuable work. Just say 'Yes!'
CAMDEN HEALTH RESEARCH PROJECT

The University of London School of Pharmacy is working in collaboration with Camden Council to investigate attitudes towards health, and how information about our genes and lifestyles can protect us from illness.

By filling in a simple questionnaire you will be contributing to important research to help the fight against cancer, heart disease and other illnesses.

The questionnaire can be emailed OR posted to you - whichever you prefer. It will take about 20 minutes to complete.

Everyone replying will be automatically entered into a prize draw to win £100!

Email sarah.carter@ulsop.ac.uk to be part of this valuable work. Just say ‘Yes!’
Dear Sir/Madam

I am a researcher from the University of London and am currently investigating attitudes towards health, illness and healthy eating.

[Your employer] has kindly agreed to distribute the attached questionnaire to you on my behalf and I would very much appreciate you taking time to complete it. It should take between 15 and 20 minutes.

PLEASE READ THE FOLLOWING INSTRUCTIONS CAREFULLY BEFORE YOU OPEN ANY ATTACHMENTS.

Please find attached two documents relating to the research project I am conducting.

The first is called 'Questionnaire'. Please open this and complete the form electronically by marking your answers to the questions with an 'X'.

Half way through the questionnaire you will be asked to open the second document titled 'Your Test Results'. Please do this when instructed and return to the questionnaire when instructed.

When you have completed the questionnaire please save the 'Questionnaire' document as a Word file and return it to me by email at this address. Please do not copy and paste it into the email. You do not have to return the document 'Your Test Results' to me.
Your data will be treated confidentially and the questionnaire is anonymous, so you do not have to give me your name. In addition, our data storage and archives comply with the Data Protection Act for anonymity and confidentiality. [Your employer] will not have access to your completed questionnaire although they may be told of the results at the end of the study.

The questionnaire has been emailed to you for convenience purposes. Should you wish to complete it on paper instead of electronically, or if you do not wish to return your questionnaire by email for any reason, please print out the questionnaire, complete it, and return to me, at the following FREEPOST address:

Sarah Carter
FREEPOST
Department of Practice & Policy
School of Pharmacy
University of London
29-39 Brunswick Square
London WC1N 1AX

If you have any queries about the research, please do not hesitate to contact me at this postal or email address, or on the telephone number 020 7753 5956.

Thank you for taking time to participate in this research - it is very much appreciated.

With kind regards,

Sarah Carter
Postgraduate Researcher

Enc.
Thank you for agreeing to complete this questionnaire. I am investigating your attitudes towards health, illness and healthy eating. Please read the questions carefully and answer as accurately as you can.

Please tick the box that best describes how you feel:

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<tr>
<th>Strongly disagree</th>
<th>Disagree</th>
<th>Neutral / Don’t know</th>
<th>Agree</th>
<th>Strongly agree</th>
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<td>If I get ill, it is my own behaviour which determines how soon I get well again.</td>
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<td>No matter what I do, if I am going to get ill, I will get ill.</td>
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<td>Having regular contact with my doctor is the best way for me to avoid illness.</td>
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<td>Most things that affect my health happen to me by accident.</td>
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<td>Whenever I don’t feel well, I should consult a medically trained professional.</td>
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<td>I am in control of my health.</td>
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<td>My family has a lot to do with my becoming ill or staying healthy.</td>
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<td>When I get ill, I am to blame.</td>
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<td>Luck plays a big part in determining how soon I will recover from an illness.</td>
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<td>Health professionals control my health.</td>
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<td>My good health is largely a matter of good fortune.</td>
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<td>The main thing which affects my health is what I myself do.</td>
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<td>If I take care of myself, I can avoid illness.</td>
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<td>Whenever I recover from an illness, it's usually because other people (for example, doctors, nurses, family, friends) have been taking good care of me.</td>
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<td>No matter what I do, I'm likely to get ill.</td>
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<td>If it's meant to be, I will stay healthy.</td>
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<td>If I take the right actions, I can stay healthy.</td>
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<td>There is nothing more important than good health</td>
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<td>Good health is only of minor importance in a happy life</td>
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<td>If you don't have your health you don't have anything</td>
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<td>There are many things I care about more than my health.</td>
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Appendices

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<td>To what extent do you believe that your diet influences your health?</td>
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<td>To what extent do you believe that your lifestyle in general influences your health?</td>
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<td>To what extent do you believe your genes influence your health?</td>
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What do you think your risk for developing heart disease is, compared with the average person? | | | | | |
Please mark on the line where you think your risk is.

How physically active would you say you are? | | | | |
Please mark on the line where you think your physical activity level is.
Appendices

How would you rate your overall health?

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</tr>
</tbody>
</table>

How many portions of fruit and vegetables do you eat, on average, per day?

<table>
<thead>
<tr>
<th></th>
<th>Less than 2</th>
<th>2 or 3</th>
<th>4</th>
<th>At least 5</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
</tbody>
</table>

How often do you eat sugary or fatty foods, like cakes, pastry or fried foods?

<table>
<thead>
<tr>
<th></th>
<th>Hardly ever</th>
<th>1 or 2 per month</th>
<th>1 or 2 per week</th>
<th>3 or 4 per week</th>
<th>Every day</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
</tbody>
</table>

Do you smoke?

<table>
<thead>
<tr>
<th></th>
<th>Yes</th>
<th>No</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>□</td>
<td>□</td>
</tr>
</tbody>
</table>

-----------------------------------------------
The following section consists of some scenarios. They are designed to measure how you would respond to threatening or risky situations.

Think carefully about each scenario, really imagining yourself in that situation. Tick all of the statements below the scenario which best describe how you would react in that particular situation.

Vividly imagine that you are afraid of the dentist and have to get some dental work done. Which of the following would you do? Tick all of the statements that might apply to you.

- I would ask the dentist what he was going to do
- I would take a tranquilizer or have a drink before going.
- I would try to think about pleasant memories.
- I would want the dentist to tell me when I would feel pain.
- I would try to sleep.
- I would watch all the dentist’s movements and listen for the sound of the drill.
- I would watch the flow of water from my mouth to see if it contained blood.
- I would do mental puzzles in my head.

Vividly imagine that you are being held hostage by a group of armed terrorists in a public building. Which of the following would you do?

- I would sit by myself and have as many daydreams as I could.
- I would stay alert and try to keep myself from falling asleep.
- I would talk with the other hostages.
- If there were a radio present I would stay near and listen to the bulletins about what the police were doing.
- I would watch every movement of my captors and keep an eye on their weapons.
- I would try and sleep as much as possible.
- I would think about how nice it’s going to be when I get home.
- I would make sure I knew where every possible exit was.
Vividly imagine that it is rumoured that several people in your department at work will be made redundant. Your manager has submitted an evaluation of your work to the Managing Director. The decision about redundancies has been made and will be announced in several days.

☐ I would talk to my colleagues to see if they knew anything about what the manager’s evaluation of me said.

☐ I would review the list of duties for my job and try to work out if I had fulfilled them all.

☐ I would go to the cinema to take my mind off things.

☐ I would try to remember any arguments or disagreements I might have had with the manager that would have lowered his/her opinion of me.

☐ I would push all thoughts of being made redundant out of my mind.

☐ I would tell friends and family that I’d rather not discuss my chances of being made redundant.

☐ I would try and think which employees in my department the manager might have thought had done the worst job.

☐ I would continue doing my work as if nothing special was happening.

Vividly imagine that you are on an aeroplane. The plane unexpectedly goes into a deep dive then suddenly levels off. The pilot announces that nothing is wrong, although the rest of the journey may be rough. You, however, are not convinced that all is well.

☐ I would carefully read the information provided about safety features in the plane and make sure I knew where the emergency exits were.

☐ I would make small talk with the passenger beside me.

☐ I would watch the end of the film, even if I had seen it before.

☐ I would call for the air steward and ask exactly what the problem was.

☐ I would order a drink from the air steward.

☐ I would listen carefully to the engines for unusual noises and would watch the crew to see if their behaviour was out of the ordinary.

☐ I would talk to the passenger beside me about what might be wrong.

☐ I would settle down and read a book or magazine.
PLEASE READ THE FOLLOWING INFORMATION CAREFULLY:

Small differences in your genes can influence how well your body metabolises foods, uses nutrients and gets rid of damaging toxins, all of which can affect your general state of health and influence your risk of developing illness.

There is a genetic test that can detect variations in the gene which influences how well the body uses the vitamin folic acid. Some people have a particular type of this gene which means that folic acid is not used in the body as well as it should be. This can increase levels of a substance called homocysteine in the blood which is linked to an increased chance of developing heart disease in the future.

People who have this type of gene can lower their risk of heart disease by increasing their dietary intake of folic acid. Folic acid is found in vegetables such as broccoli, Brussels sprouts, cabbage and cauliflower. It is also found in smaller amounts in some breakfast cereals, bread, eggs, lentils and peanuts.

Now imagine that you have taken this genetic test to find out which variation of the gene you have.

Now open the envelope to find out your results.
*COMPLETE THE REMAINING QUESTIONS AFTER YOU HAVE OPENED YOUR TEST RESULTS*

Continue to imagine that you have received this information about your genes.

Please tick the box which best describes how you feel after receiving this test result:

<table>
<thead>
<tr>
<th>At present I feel...</th>
<th>Not at all</th>
<th>Somewhat</th>
<th>Moderately</th>
<th>Very much</th>
</tr>
</thead>
<tbody>
<tr>
<td>...calm</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>...tense</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>...upset</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>...relaxed</td>
<td></td>
<td></td>
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</tr>
<tr>
<td>...content</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>...worried</td>
<td></td>
<td></td>
<td></td>
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</tbody>
</table>

How certain are you that you could manage to eat a healthy diet...

<table>
<thead>
<tr>
<th>Even if you need a long time to develop the necessary routines</th>
<th>Very uncertain</th>
<th>Rather uncertain</th>
<th>Rather certain</th>
<th>Very certain</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Even if you have to try several times until it works</th>
<th></th>
<th></th>
<th></th>
<th></th>
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</thead>
</table>

<table>
<thead>
<tr>
<th>Even if you have to rethink your entire way of nutrition</th>
<th></th>
<th></th>
<th></th>
<th></th>
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</thead>
</table>

<table>
<thead>
<tr>
<th>Even if you do not receive a great deal of support from others when making my first attempts</th>
<th></th>
<th></th>
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</thead>
</table>

| Even if you have to make a detailed plan.                                                      |                |                  |                |              |

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Appendices

<table>
<thead>
<tr>
<th>Not at all</th>
<th>Not much</th>
<th>A little</th>
<th>A lot</th>
<th>Very much</th>
</tr>
</thead>
</table>

To what extent do you believe that your diet influences your health?

To what extent do you believe that your lifestyle in general influences your health?

To what extent do you believe your genes influence your health?

---

*Continue to imagine that you have received this information about your genes.*

Now that you have been given this genetic information, what do you think your risk for developing heart disease is, compared with the average person?

<table>
<thead>
<tr>
<th>Very low</th>
<th>Average</th>
<th>Very high</th>
</tr>
</thead>
</table>

Please mark on the line where you think your risk is.

<table>
<thead>
<tr>
<th>Very unlikely</th>
<th>Very likely</th>
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</thead>
</table>

How likely is it that you will change your diet to help lower the risk of heart disease?

Please mark on the line where you think your likelihood is.

---
The following questions are about the use of genetic information. Please tick the box under the answer which best describes how you feel:

If you had actually taken this genetic test, how worried would you be that your genetic information would be used by other companies or by the government?

<table>
<thead>
<tr>
<th>Not at all</th>
<th>A little</th>
<th>Moderately</th>
<th>Quite a lot</th>
<th>Extremely</th>
</tr>
</thead>
<tbody>
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If this genetic test were to come onto the market, how worried would you be that insurance companies would use your genetic information to calculate insurance premiums?

<table>
<thead>
<tr>
<th>Not at all</th>
<th>A little</th>
<th>Moderately</th>
<th>Quite a lot</th>
<th>Extremely</th>
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</thead>
<tbody>
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</table>

If this genetic test were to come onto the market, how worried would you be that employers would use your genetic information to influence their decisions about offering you a job or a promotion?

<table>
<thead>
<tr>
<th>Not at all</th>
<th>A little</th>
<th>Moderately</th>
<th>Quite a lot</th>
<th>Extremely</th>
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How well did you understand the information you were given about the genetic test and your results?

<table>
<thead>
<tr>
<th>Not at all</th>
<th>A little</th>
<th>Moderately</th>
<th>Quite a lot</th>
<th>Extremely</th>
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The following questions are about you. Please tick the box under the answer which describes you best:

What is your age?

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<thead>
<tr>
<th>Age Range</th>
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<td>18-20</td>
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<td>26-30</td>
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<td>41-45</td>
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</tbody>
</table>

Are you...?

Male □ Female □

Are you...?

Single □ Married □ Cohabitating □ Divorced □ Widowed □

What ethnicity are you?

White □ Indian □ Pakistani or Bangladeshi □ Black Caribbean □

Black African □ Chinese □ Mixed □ Other □

What is your highest qualification?

None □ GCSE □ A level □ Higher education □ Degree or equivalent □

Other eg. Postgraduate (please specify)...

Thank you very much for your time. Please feel free to contact me at any time should you have any questions.

Sarah Carter
020 7753 5956
sarah.carter@ulsop.ac.uk