The Informational Aspects of Direct-to-Consumer Genetic Tests

By

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Abstract

Background Direct-to-consumer (DTC) genetic tests are tests sold directly to consumers, normally without the involvement of healthcare professionals, which aim to provide consumers with their relative genetic risk for various complex diseases. Providers claim that this information will enable and encourage consumers to improve their health behaviour in order to reduce their likelihood of contracting diseases for which they are at an increased genetic risk. However, there are many criticisms and concerns about DTC genetic tests in the literature. Two common concerns are the lack of positive effects, and possible negative effects, that the information generated by the tests may have on consumers' health behaviour and health anxiety, and the identified poor quality of information provision on the websites of providers of DTC genetic tests. Although the literature contains some research in these areas it is noticeably limited and occasionally contradictory.

Aim and Methods The aim of the research was to investigate the informational aspects of direct-to-consumer genetic tests, including the provision of information by the companies, consumers' information needs and information-seeking behaviour and the effect of the information generated by the tests on health behaviour and health anxiety. The research consisted of three studies: a survey of 275 consumers and potential consumers of DTC genetic tests, in-depth email interviews with 36 consumers of DTC genetic tests and a content analysis of the information provided on all identified providers' websites.

Results Positive or neutral changes in health behaviour were identified in a large minority of respondents who had been exposed to genetic risk information, along with the mechanisms by which the information prompted or contributed to change. A minority reported a change in health anxiety, mainly but not exclusively a decrease, with mechanisms again identified. Consumers reported a wide variety of information needs, the most common of which were information to do with the coverage and accuracy of the tests. The provision of information on providers' websites varied considerably, both between and within providers, but was generally poor. However, most consumers used other sources alongside these websites, the most common of which was blogs.

Conclusions The results suggest that concerns about possible negative effects of the information generated by the tests are unfounded and that a large minority of consumers have improved health behaviour and decreased health anxiety after purchase. The results also suggest that concern about information provision on providers' websites is justified; although this is mitigated by consumers' general use of other sources alongside the websites, it is likely that a substantial number of consumers do not have access to enough information to give fully informed consent to the test.

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Table of Contents

1 Introduction

1.1 Background

The modern field of genetics can be traced back to the 19th century, when an Austrian monk named Gregor Mendel experimented with the heredity properties of pea plants (Jorde et al 2006, pp.1, 3; Sturtevant 1965, p.10). Mendel noticed that the plants passed on physical characteristics to their offspring in precise ratios, from which he concluded that each plant had two pieces of hereditary information for each characteristic (DeSalle & Yudell 2004, pp.10-12).

By the turn of the twentieth century chromosomes had been discovered, and it had been correctly suggested that these were the vectors for "hereditary material" (Sturtevant 1965, pp.18-19). In 1944 Avery, McCarty and Macleod discovered that this hereditary material was in the form of deoxyribonucleic acid (DNA) (DeSalle & Yudell 2004, p.20), and in 1953 Watson and Crick famously discovered its double helix structure (DeSalle & Yudell 2004, pp.21-22; Martini & Bartholomew 2007, p.47)

In the 1970s a major breakthrough occurred when a method was created for determining the order of the nucleotide bases that make up a DNA strand. Although a slow process, this was the first time it had been possible to unravel, or sequence, a part of the genetic code (DeSalle & Yudell 2004, p.40). In 1990 the Human Genome Project began with the aim of sequencing the entire human genetic code, or genome, and in 2003 it succeeded in producing a sequenced genome of three billion nucleotide bases (DeSalle & Yudell 2004, p.48; Jorde et al 2006, p.3).

Genetics, and the new field of genomics, have many benefits, from proving identity (Debenhem 1990, pp.38-40) to furthering the understanding of evolution (DeSalle & Yudell 2004, pp.133-135). However, the benefits with regard to health are potentially revolutionary. For example, it is currently possible to test an individual's genetic code for many different diseases caused by genetic disorders (Jorde et al 2006, p.278). Newborns are screened for phenlyketonuria, a genetic disease with severe symptoms that are easily preventable if caught at an early age (Scriver 2007). At-risk, but healthy, individuals can be tested to ensure they do not carry single copies of genes that, if shared with their partner, could cause diseases such as Tay-Sachs or Thalassemia in their children (Fuhrmann & Vogel 1983, pp.36-37; Jorde et al 2006, p.282; Zlotogora 2009). These commonly-used techniques may in the future be joined by others currently under development including the use of gene therapy to cure certain diseases (Jorde et al 2006, pp. 296-301), the tailoring of an individual's medicine to their genome (DeSalle & Yudell 2004, pp.130-132; Primrose & Twyman 2004, pp. 109-

110) and the use of an individual's genome to predict their susceptibility to many different types of complex disease (DeSalle & Yudell 2004, p. 129).

These potential benefits have spurred a rapid advance in genomics; one that has been witnessed during this PhD. From a price of three billion dollars for the first genome sequence in 2003 (Robertson 2003), the price had dropped to 48 thousand dollars by 2010 (Los Angeles Times 2010). In 2013 the price has dropped even further, to an incredibly low five thousand dollars.[1] At the outset of this PhD only a handful of full human genomes had been sequenced, but now the 1,000 genome project alone contains over 1,000 (1000 genomes, 2013), and an organisation such as the National Health Service (NHS) can plan to sequence 100,000 within the next five years (gov.uk 2012).

The potential benefits of genomics have also led to the creation of direct-to-consumer (DTC) genetic tests. DTC genetic tests are tests that an individual can purchase from a website without the involvement of a healthcare professional, and that claim to provide a personalised genetic risk assessment for a large number of complex diseases (Bloss et al. 2011a; Samuel et al. 2010). The aim of providers is to encourage consumers to modify their health behaviour in order to reduce their chances of contracting diseases for which they are at an increased risk (cf. 23andMe 2013a; Inherent Health 2009). Although prediction of disease risk is a potential benefit of genomics, as mentioned above, the general consensus is that it is currently not possible to do this accurately. Therefore, there are many people who are concerned that DTC genetic tests may be inaccurate (cf. Murray et al 2010).

Aside from genetics and medicine, DTC genetic tests raise issues in a broad range of disciplines. Some of the most important of these issues are the informational aspects of the tests, such as the provision of complex genetic information to consumers and their reaction to the large volumes of information generated by the tests. It is on these issues that this thesis is focused.

1.2 Research Need

A review of the literature highlighted many concerns about DTC genetic tests. Two of the most commonly mentioned are the effect that the information generated by the tests may have on consumers' health behaviour and health anxiety and the poor quality of information provision, with regard to informed consent and understanding of the tests, on providers' websites. Despite a

[1] Since submission of this thesis a company named Illumina have released a machine that is claimed to have the ability to sequence an entire human genome for under one thousand dollars (Illumina 2014).

reasonable quantity of research into DTC genetic tests in recent years, these are still areas in which large gaps remain in the literature.

When the research aim, questions and objectives were first formulated no published research into the effects of the information generated by DTC genetic tests on health behaviour and anxiety could be found. Despite numerous opinion pieces about the possible negative effects of this information, or at the very least the lack of a positive effect, the sometimes vociferous comments were based only on conjecture, not evidence. Although several studies have been published since, to the author's knowledge there are still only three studies that have examined the effects of the information provided by DTC genetic tests for multiple disease risk assessments on the health behaviour and/or health anxiety of a sufficiently large group of participants: Gordon et al (2012), Kaufman et al (2012) and Bloss et al (2011b). However, these studies are far from conclusive and all have potential biases. Firstly, the results are contradictory. Although Gordon et al (2012) and Kaufman et al (2012) found that receipt of test results had caused changes to the health behaviours of a minority of participants, Bloss et al found no such changes. Secondly, in the studies conducted by Bloss et al (2011b) and Gordon et al (2012), participants were not actual consumers of DTC genetic tests, but participants who had been given a free or subsidised test as part of the study; their results therefore may not be generalizable to ordinary consumers of a relatively expensive new product. Although the study by Kaufman et al (2012) was based on a survey of actual consumers, which negates this problem, respondents were contacted through the providers of the tests and so independence cannot be guaranteed. Finally, the studies by Gordon et al (2012) and Kaufman et al (2012) both relied solely on enquiring if participants' health behaviour had changed as a result of receiving their genetic risk information. Although this did provide useful information, there is an inherent possibility of bias in this method, such as incorrect recall or demand characteristics bias. Although Bloss et al (2011b) measured participants' health behaviours before and after testing, and hence removed the possibility of these biases, they only analysed two behaviours (fat intake and exercise); any changes to other health behaviours would therefore have been missed. As Bloss et al (2011a, p.132) state, more research in this area is "desperately needed".

There are a reasonable number of well designed studies that have examined the information provision on DTC genetic test providers' websites, as described in section 2.4.2.2. These studies have generally been content analyses, with information provision compared against either professional recommendations for the information that should be provided or criteria created by the researchers themselves. These studies have unanimously shown that information provision is generally less than acceptable. However, to the author's knowledge, no study has examined the

provision of information from the consumers' viewpoint. For example, no research has been found that has investigated the information that consumers themselves wish to know and whether or not this information is provided on the websites. Also missing is research into how consumers search for information; whether they are wholly reliant on providers' websites or whether they search for information themselves. Finally, no study has been found that has investigated consumers' opinions of the information provided to them.

1.3 Research Aim, Questions and Objectives

1.3.1 Aim

The aim of the research was to investigate the informational aspects of direct-to-consumer genetic tests, including the provision of information by the companies, consumers' information needs and information-seeking behaviour and the effect of the information generated by the tests on health behaviour and health anxiety.

1.3.2 Objectives

The research had the following 14 objectives:

1. To identify a sample of consumers of DTC genetic tests and a sample of individuals who are interested in the tests but have not yet purchased one.
2. To inquire into changes to consumers' health behaviour and health anxiety after receipt of DTC genetic test result information.
3. To compare the current health behaviour and health anxiety of consumers of DTC genetic tests with individuals who are interested in purchasing a test or who have purchased one but not yet received their results.
4. To assess the mechanisms through which the information from a DTC genetic test can affect health behaviour and health anxiety.
5. To assess the information needs of consumers of DTC genetic tests.
6. To assess the information-seeking behaviours of consumers of DTC genetic tests.
7. To assess changes to consumers' information needs after receipt of DTC genetic test result information.
8. To assess changes to consumers' information-seeking behaviours after receipt of DTC genetic test result information.
9. To identify all providers of DTC genetic tests for multiple disease risk assessment.
10. To identify recommendations for the information that should be provided by providers of DTC genetic tests.

11. To analyse the information provided on the websites of providers of DTC genetic tests with regard to recommendations of information that should be provided and consumers' self-identified information need.
12. To assess consumers' opinions about DTC genetic tests.
13. To assess consumers' experiences with DTC genetic tests.
14. To develop a model or models that describe the research findings regarding consumers' experiences when purchasing a DTC genetic test and their related information-seeking behaviour.

1.3.3 Research Questions

The 14 objectives arose from the following research questions:

1. What effect does the information from a DTC genetic test have on consumers' health behaviour and health anxiety?
2. How does the information from a DTC genetic test affect consumers' health behaviour and health anxiety?
3. What are consumers' information needs and information-seeking behaviours?
4. What effect does the information from a DTC genetic test have on consumers' information needs and information-seeking behaviours?
5. Are consumers' information needs met by the information provided on the websites of companies that sell DTC genetic tests?
6. What are consumers' opinions about, and experiences with, DTC genetic tests?

1.4 Outline of Thesis

The introduction has given a basic background of the research area, the justification for the research and the research aim, objectives and questions. This is followed by Chapters 2 and 3 that examine the research area in detail. Chapter 2 focuses on genetics, genomics and genetic testing. It describes their history, the current situation and possible future benefits, and reviews the published research on DTC genetic tests. Chapter 3 describes information behaviour research and health information, and examines a selection of information behaviour models.

Chapter 4 describes the Research Methodology. This chapter begins by describing the theoretical framework of the research and the overall research design. It then describes the methods for the three studies conducted during the course of the research: a survey, email interviews and a content analysis.

Chapters 5, 6 and 7 present the findings of the survey, email interviews and content analysis. Each study is the focus of a single chapter, which presents the study's results, the analysis of the results and a discussion of the findings in isolation. Chapter 8, Final Discussion, then brings the most important findings together and examines the results as a whole, comparing them to each other and to the literature. It assesses the fulfilment of the aim and objectives, presents an analysis of the ethical implications of the findings and two models of the results.

The final chapter of the thesis, Chapter 9, is the Conclusions. This begins by identifying the contributions of the research, discusses its wider implications, describes its limitations and identifies further research to be conducted in the area. It finishes with a final conclusion of the thesis.

2 Background and Review of Genetics, Genomics, and Genetic Tests

2.1 Introduction

The aim, objectives and research questions of this thesis, as shown in the previous chapter, have a large scope and touch on a wide range of issues. As such, it is necessary for a number of topics to be examined.

The first section of the chapter provides a background on genetics and genomics. It gives an overview of its history, the current state of knowledge and the uses and potential future uses of genetic information. This section is important as it gives a context to the research and introduces the potential advantages and disadvantages of genetic testing.

The second section describes the ethical and legal issues that relate to genetics. Again, this section provides a necessary context for the research; many of the concerns about DTC genetic tests, both professionals' and consumers', relate to these issues.

The third and final section of this chapter focuses on DTC genetic tests. It begins by describing the tests, highlights common criticisms and concerns about them and describes the position regarding their legality. Finally, it examines the published research into DTC genetic tests.

2.2 Genetics

2.2.1 Background

2.2.1.1 The History of Genetics

DeSalle and Yudell (2004, p.4) describe genetics as "the study of the mechanisms of heredity". Heredity is the passing of traits from an organism to its descendants (DeSalle & Yudell 2004, p.5) and theories about the mechanisms of it go back at least as far as Hippocrates (Sturtevant 1965, p.1). The utilisation of the process of heredity goes back even further, to the domestication of plants and animals many thousands of years ago (Stubbe 1972, pp.1-9). However, the modern theory of genetics can be traced back to a 19th century Austrian monk named Gregor Mendel (Jorde et al 2006, pp.1,3).

Mendel published a paper in 1866 which detailed experiments that he had performed with pea plants, and the conclusions he had drawn from them (Sturtevant 1965, p.10). He looked at characteristics of the plants that had two different traits. For example, the seeds were either yellow or green, and the stems were either long or short. After cross-breeding and self-fertilizing many

thousands of plants, Mendel realized that these traits were passed to the offspring in very precise ratios (for example, in the second generation three quarters of the plants consistently had one of the traits, and a quarter the other), the traits for each characteristic were passed on independently of each other (for example, the trait that was passed on for the height of the plant was not affected by which trait was passed on for the colour of the seed) and that breeding two plants with one trait could sometimes produce offspring with the other trait (for example, two plants with yellow seeds could have offspring with green seeds). Mendel concluded from these results that each plant contained two pieces of hereditary information for each characteristic, which he called "form factors", that each parent passes one of these to their offspring (with an equal chance of which one is passed on), and that each trait was passed on independently from the other traits. He also concluded that some of the factors were dominant and some recessive; dominant factors would always determine the trait if there was at least one of them and recessive factors would only determine the trait if there were two of them (and hence no dominant factors) (DeSalle & Yudell 2004, pp.10-12). In modern terminology, Mendel realised that each parent had two alleles (or variants) for each gene, only one of which would be passed on to the offspring from each parent and the phenotype (or characteristics) of the offspring would be determined by which alleles had been passed on, with a dominant allele taking precedence over a recessive allele.

It is now recognised that the hereditary process is more complicated than that described by Mendel. For example, many traits are caused by more than one gene (polygenic traits) or by a combination of multiple genes and the environment (multifactorial traits) (although each of the genes in a multiple gene trait is still inherited in the way Mendel suggested) (Jorde et al 2006, p.248), some alleles are co-dominant, resulting in a phenotype which includes the traits of both of the alleles (Martini 2006, p.1100) and some genes are located close enough on the same chromosome that they are usually passed on together (a situation called linkage) (Jorde et al 2006, pp.161-162). However, apart from these complications, Mendel's conclusions are still considered to be accurate and Mendel's laws are still taught in genetics classes.

Mendel's paper did not receive much recognition until the year 1900, when three other researchers suggested similar theories of inheritance (Sturtevant 1965, pp.25-30; Jorde et al 2006, p.12). By this time chromosomes had been discovered, as well as the halving of the number of chromosomes in meiosis (the process that produces egg and sperm cells and results in only one allele for each gene being passed on to the offspring per parent) and the idea of chromosomes carrying "hereditary material" had been proposed (Sturtevant 1965, pp.18-19).

In 1944, Avery, McCarty and Macleod performed an experiment where they mixed benign bacteria with similar bacteria (the same species) that were virulent but dead. They found that the benign bacteria became virulent; therefore the virulent bacteria were passing on traits to the benign. Although there was no effect on the benign bacteria if they were only mixed with proteins which had been isolated from the virulent bacteria, when they were mixed with the virulent bacteria's nucleotides (which make up DNA) they became virulent. This showed that DNA was the hereditary material (DeSalle & Yudell 2004, p.20) (although it should be noted that the hereditary material in a small number of organisms is a closely related chemical called ribonucleic acid (RNA) (Dawkins 2010, pp.420-421)).

In 1953, the structure of the chains of DNA that store hereditary information was worked out by Watson and Crick (DeSalle & Yudell 2004, pp.21-22). They described it as consisting of "two helical chains coiled round the same axis" (Watson & Crick 1953, p.737); described now as a double helix (Martini & Bartholomew 2007, p.47). This was perhaps the most famous discovery in genetics history; however, it should be noted that the inspiration for this discovery came after Watson was shown an X-ray photograph of DNA crystals, without the knowledge of the person who had taken them. This person was a chemist named Rosalind Franklin, whom Crick later acknowledged was probably very close to coming up with the answer herself (DeSalle & Yudell 2004, pp.22-26).

After the discovery of the structure of DNA came the discovery of how DNA passes on hereditary information. Proteins perform many functions in the body, such as digesting food and transmitting nerve impulses. They are composed of long chains of amino acids, the order of which are determined by the genetic code contained within DNA. Watson and Crick (with the help of a letter from a physicist named George Gamow) determined that there are 20 different amino acids that can be used to make proteins. It was soon realised that three nucleotides (the components of DNA) code for one amino acid, and in 1961 Matthaei and Nirenberg discovered which nucleotides make up the code for an amino acid named phenylalanine. By 1965 the codes for all twenty of the amino acids had been worked out (DeSalle & Yudell 2004, pp.28-34).

In 1975 Edward Southern invented the Southern blot. This is a technique which uses agarose gel and an electric current to separate DNA molecules and (with the use of radioactive gene fragments) allows genes of interest to be detected (DeSalle & Yudell 2004, pp.37-39). This can be used to find variations in genes between individuals (Jorde et al 2006, p.43).

In the 1970s two groups simultaneously discovered a way to determine the order of the nucleotides in a DNA strand; a process named sequencing. One group was based at Harvard and headed by

Walter Gilbert, the other was based at Cambridge and headed by Frederick Sanger. Their technique involved copying a piece of DNA and using a chain terminator to interrupt it at various points. These chain terminators interrupt the copying of the DNA at specific nucleotides in the chain, and if they are radioactively labelled (and then the DNA separated with gel and an electric current) the position of the different nucleotides in the sequence can be identified. However, it should be noted that this is a very slow process (DeSalle & Yudell 2004, p.40).

In 1983 a scientist named Kerry Mullis came up with an idea to enable large quantities of DNA to be quickly replicated. The process is called the polymerase chain reaction and can make billions of copies of short DNA sequences. The process is in the form of a continuous cycle, whereby a solution containing the DNA to be copied, unattached nucleotides, two primers which attach to the DNA next to the part of the sequence that will be copied and an enzyme called DNA polymerase is heated to 94-95 degrees centigrade, cooled to 35-65 degrees, heated to 70-75 degrees and then heated again to 94-95 degrees and so on. This causes the DNA to denature (the strands separate), the primers to attach, the replication to occur and then the DNA (both the old and the new strands) to denature again etc (Jorde et al 2006, pp.48-49; DeSalle & Yudell 2004, p.41).

By 2003, the first complete human genome was fully sequenced by the Human Genome Project (Jorde et al 2006, p.3). This is discussed further in section 2.2.2.1.

2.2.1.2 Genetic Structure and Function

Genes are in the form of a chemical called deoxyribonucleic acid (DNA) (Jorde et al 2006, p.6). In the cells of most organisms, the DNA is organised into two strands which are twisted into a double helix shape, which is itself coiled up into chromosomes that are stored in the nucleus of the cell (Martini & Bartholomew 2007, pp.47, 76-77). All DNA molecules are composed of a phosphate group, deoxyribose and a pentose sugar. They also all have a nitrogenous base, which comes in four different types: cytosine (C), thymine (T), guanine (G) and adenine (A). When DNA is in a double helix, the bases point inwards and bind to each other (Jorde et al 2006, pp.6-7). 'A' always bonds with 'T' and 'C' always bonds with 'G', which means that the two strands are complementary (DeSalle & Yudell 2004, pp.24-25).

Genes work by controlling the construction of all of the proteins used by organisms (Jorde et al 2006, p.6). Since, as described earlier, proteins carry out a large range of functions in the body (DeSalle & Yudell 2004, p.29) this allows genes to "influence all aspects of body structure and function" (Jorde et al 2006, p.6). Proteins are made of long chains of amino acids, and it is the type and order of these amino acids that genes code for. The genetic code is the order of the four different DNA bases

on a DNA strand, with every three bases (in a gene coding region) coding for an amino acid. For example, three T's in a row codes for the amino acid phenylalanine. As there are 64 ways that these sequences of three bases can be arranged and just 20 different amino acids, most amino acids can be coded for by more than one sequence of three bases; there are also sequences of three bases that code for starting and stopping protein synthesis (DeSalle & Yudell 2004, pp.28-34).

2.2.1.3 Genetic Diseases

Genetic diseases can be the result of four different types of genetic disorders. These are single-gene disorders, multifactorial disorders, chromosome disorders and mitochondrial disorders (Jorde et al 2006, p.3).

2.2.1.3.1 Single-gene Disorders

Single-gene disorders are disorders where a disease is caused by an alteration in a single gene. These alterations can be caused by many different types of mutations that change the genetic code and cause a different arrangement of amino acids in a protein. Single-gene disorders are also known as Mendelian conditions, as they follow a pattern of Mendelian inheritance (Jorde et al 2006, pp.3, 29-31). These conditions can either be the result of mutated genes passing down through families, or the result of a new mutation in the affected person (occurring in either the egg or sperm cell which combined to make the embryo) (Fuhrmann & Vogel 1983, pp.18-26).

There are a very large number of single-gene disorders (Jorde et al 2006, pp.3-4). One example is Huntington's disease. This is a terminal disease that is characterised by involuntary movements and chorea. It was discovered that it is caused by a sequence of three bases repeating itself a large number of times in the gene coding for Huntingtin protein (the HD gene), causing extra amino acids to be added to the protein (Gusella & MacDonald 2009).

2.2.1.3.2 Multifactorial Disorders

Multifactorial disorders are "caused by interactions between genetic predispositions and environmental factors" (Penchaszadeh 2001, p.310), with more than one of each of these factors usually involved (Jorde et al 2006, p.3; Twyman 2003). This means that they have a much lower level of heritability than single-gene disorders (Twyman 2003) and do not follow a pattern of Mendelian inheritance (although the genes involved will do individually) (Jorde et al 2006, p.248).

One example of a multifactorial disorder is coronary artery disease (CAD). Although occasionally a single-gene disorder, CAD is usually a multifactorial disorder. The environmental factors which increase the chance of CAD are well known, and include smoking, a high fat consumption and being physically inactive. The genetic factors are much less certain however, although it is estimated that

"the overall genetic contribution toward the development of CAD ... range[s] from 20% to 60%". Genes whose variants may increase the risk of CAD include Apolipoprotein E, Lipoprotein lipase and Apolipoprotein B (Nordlie et al 2005, p.668).

2.2.1.3.3 Chromosome Disorders

In a chromosome disorder a whole chromosome or a segment of one is either altered, duplicated or completely missing (Jorde et al 2006, p.3). Unlike the other types of genetic disease, chromosome disorders are not hereditary, although a susceptibility to them may be (Primrose & Twyman 2004, p.90).

An example of a chromosome disorder is Down syndrome (Jorde et al 2006, p.3). Down syndrome is caused by the individual having three copies of chromosome 21 rather than the ordinary two copies. It is unclear how this causes Down syndrome, although there are suggestions that the genes on the extra chromosome may interact with the rest of the genome (Neri & Opitz 2009).

2.2.1.3.4 Mitochondrial Disorders

Mitochondria are organelles inside cells in which chemical reactions occur that provide most of a cell's energy (Martini & Bartholomew 2007, pp.74-75). Mitochondria contain their own DNA which is separate from the rest of the genome; it is stored in a circular shape and is passed on solely from the mother. Mutations in this DNA can cause genetic diseases (Jorde et al 2006, pp.101, 103-105).

An example of a mitochondrial disorder is Leber hereditary optic neuropathy, which involves rapid central field vision loss (Jorde et al 2006, p.105).

2.2.1.4 Single Nucleotide Polymorphisms

Single nucleotide polymorphisms (SNPs) are common variations that occur at a single nucleotide on a DNA sequence. They occur approximately once in every thousand bases (Gardner & Davies 2009, p. 173; Jorde et al 2010, p.41) and "are the most common type of variation in the human genome" (Jorde et al 2010, p.43). Some SNPs can cause genetic disease but the majority do not (Jorde et al 2010, pp.41-43).

Due to the profusion of SNPs in the genome they are of particular use as "genetic markers" (Gardner & Davies 2009, p. 173). Case-control studies can be used to determine if the presence of an SNP is associated with a particular disease, which, if so, is indicative that the SNP is within or near to the gene responsible. This is the premise of genome-wide association studies (GWAS) which can determine the presence of SNPs at many thousands of locations across the genome (Korf & Irons 2013, pp.92-93; Jorde et al 2010, p.163). SNPs are also often used as genetic markers by providers

of DTC genetic tests (described in section 2.4) which can, as in GWAS, sequence the location of many thousands of SNPs in one go (Korf & Irons 2013, pp.214-215).

2.2.1.5 Epigenetics

Epigenetics describes "changes in gene expression" that are not caused by changes to the actual sequence of DNA bases (Gardner & Davies 2009, p.30) but are still inherited by each descendent of the cell in which they originally occurr (Korf & Irons 2013, p.16). Epigenetic changes involve the deactivation of genes by chemical modification of DNA (Korf & Irons 2013, p.16) which can occur through either DNA methylation, RNA-associated silencing or histone modification (Gardner & Davies 2009, p.30). Although these modifications result in genes that are "permanently silenced" Korf & Irons 2013, p.16), it is considered that the modifications are "potentially reversible" (Gardner & Davies 2009, p.30).

Epigenetics may be used by the body as a method of "fine control of gene expression" (Gardner & Davies 2009, p.30). However, epigenetic changes can lead to health problems; it has been demonstrated, for example, that epigenetic changes are an important mechanism in the development of tumours in many different types of cancer (Al-Chalaby & Almasy 2009, pp.149-150). Also, although a relatively new field, epigenetic research is beginning to reveal associations between epigenetics, the environment and susceptibility to disease. For example, susceptibility to type two diabetes may be influenced by epigenetic changes caused by differences in foetal nutrition (Korf & Irons 2013, p.18). In patients with Borderline Personality Disorder who have a history of abuse in childhood, epigenetic changes to the gluticord receptor gene have been significantly correlated with both severity of abuse and severity of the disorder, suggesting an association between the two (Martin-Blanco et al 2014). There is evidence that epigenetics may affect susceptibility to pain and may influence the healing process (Lirk et al 2014) and epigenetic changes may explain the increase in susceptibility to prostate cancer with increasing age (Damaschke et al 2013).

2.2.1.6 Benefits of Genetics

The study of genetics has many benefits, both medical and non-medical.

2.2.1.6.1 Genetic Diseases

Some of the main medical benefits of genetics are to do with genetic diseases.

2.2.1.6.1.1 Genetic Testing

One of the main ways that genetics can help against genetic diseases is through genetic testing. Genetic testing involves testing for genetic disorders that may cause a disease; this can be carried

out either by directly analysing the genetic material or by analysing the proteins created by it. Genetic testing can either involve population screening, where all members of an at-risk population are tested for genetic disorders, or the genetic testing of a family that has, or has had, members with a specific genetic disorder (Jorde et al 2006, p.278).

An example of population screening is testing for a disease called phenylketonuria (PKU) in newborns. PKU is characterised by impairment in cognitive development and function and involves a disorder of the phenylalanine hydroxylase gene. This disrupts the breakdown of an amino acid named L-phenylalanine, the build-up of which causes the symptoms of PKU. It has been found that a diet low in this amino acid prevents the symptoms from occurring, and since there is "a simple laboratory test" to test for PKU, there are now large numbers of newborn screening programs around the world (Scriver 2007, p.832).

A second type of population screening is the screening of heterozygotes for genetic diseases that are caused by a recessive allele (Jorde et al 2006, p.278). Since these heterozygous carriers would not suffer from the disease, they may not know that they carry a disease-causing allele. However, if they have children with another heterozygous carrier, one or more of their children may inherit both disease-causing alleles and develop the disease (Fuhrmann & Vogel 1983, pp.36-37). In order to prevent this happening by chance, the partners of heterozygotes can be tested to make sure that they are themselves not heterozygous (Fuhrmann & Vogel 1983, p.42), and populations that have a high frequency of a disease allele can be the focus of population screening efforts (Jorde et al 2006, pp.281-282). Examples of these screening programs include the screening for Tay-Sachs disease amongst Ashkenazi Jews (Jorde et al 2006, p.282) and for Thalassemia in Greek Cyprus (Zlotogora 2009). This screening allows potential heterozygous parents to make informed decisions about whether or not they want to have children, and allows them to consider the full range of options such as adoption and pre-implantation diagnosis (Zlotogora 2009). If two heterozygous adults do decide to have children, the genetic screening can make it clear that the child needs to be closely monitored (Fuhrmann & Vogel 1983, p.44).

Prenatal diagnosis is a third type of screening. This may be offered for various reasons, such as if both parents are heterozygous carriers of the same recessive disease causing allele (Jorde et al 2006, pp.281-282) or to test for Down syndrome if the mother is over 35 (Martini & Bartholomew 2007, p.669). This is a controversial type of screening for several reasons. Firstly, the current prenatal diagnosis technique can harm both the mother and the foetus (Martini & Bartholomew 2007, p.669). Secondly, if it is detected that the foetus has a serious genetic disease it often leads to termination of the pregnancy. The ethics of this situation are the subject of a lot of debate (Newson 2008). Also,

there are arguments as to whether or not prenatal diagnosis stigmatises people who have the genetic disease (Newson 2008).

A fourth type of screening is presymptomatic diagnosis. This is the genetic testing of at-risk individuals (e.g. because of a family history of a disease) for mutations that will cause a genetic disease before they develop any symptoms. This has both advantages and disadvantages (Jorde et al 2006, pp.282,284). For example, Huntington disease is a fatal disease that may not become noticeable until middle age (Gusella & MacDonald 2009). It is caused by a dominant allele, which means that the children of someone with the disease have a 50% chance of suffering from it themselves (Jorde et al 2006, pp.69,71). In this situation, the advantages of testing are that it can help the individual to make decisions about family planning and that it can reduce the anxiety of 'not knowing'. However, the disadvantages are that individuals who know that they have the disease also know that each of their children has a 50% chance of inheriting it, and that they themselves have a disease which currently has no cure (Hines et al 2010). This type of testing can be worthwhile though; in the case of some disorders, such as a dominant mutation which can cause breast cancer, this type of genetic testing can make "early diagnosis and treatment" more likely (Jorde et al 2006, p.282) and hence increase the survival rate.

2.2.1.6.1.2 Gene Therapy

Gene therapy is an idea that is still in its early stages. The basic idea is to alter the expression of genes in an individual's somatic cells as a treatment for disease. Target diseases include both inherited genetic disorders such as Haemophilia and non-inherited complex diseases such as AIDS. Gene therapy is comprised of two main approaches: gene replacement therapy and gene blocking therapy. Gene replacement therapy involves the addition of a gene into the DNA of an individual's somatic cells in order to induce synthesis of a particular protein. This can be achieved through the use of various techniques such as cell fusion, electroporation and viral vectors and is useful where the presence of a protein may help to treat a condition, such as if an individual has inherited a genetic mutation which prevents them from producing a particular protein or where a tumour suppressor gene has been inactivated in cancer cells. In contrast, gene blocking therapy aims to prevent synthesis of a protein that, due to a mutation, causes damage to the body. Gene blocking techniques generally focus on causing interference to a cell's messenger RNA, a chemical that plays an important role in the process of constructing proteins from the genetic code. One technique used to achieve this is the creation of a strand of DNA to bind with the messenger RNA in order to prevent its involvement in protein synthesis. Another is to engineer a ribosome (an enzyme in the cell) in order to destroy specific sequences of messenger RNA (Jorde et al 2010, pp.273-280). In recent years there has been much research into gene therapy with mixed success. For example, an

attempt to correct a T-cell deficiency in patients with x-linked severe combined immunodeficiency resulted in almost complete mitigation of the deficiency for 17 out of 20 participants. However, only half of participants were able to cease treatment with immunoglobulin replacement, five participants developed T-cell leukaemia and, as a result, one participant died (Nienhuis 2013). Gene therapy has been used as a possible treatment for a form of hereditary blindness named Leber congenital amaurosis which is caused by both dysfunction and degeneration of photoreceptors. Although a substantial improvement was observed in the dysfunction of photoreceptors there was no improvement in their state of degeneration (Cideciyan et al 2013). A recent trial has found an improvement in muscle control in patients with Parkinson's disease who injected with a retroviral vector in order to induce brain cells that do not normally produce dopamine to do so (Palfi et al 2014).

2.2.1.6.2 Proving Identity

One non-medical benefit of genetics is that it can be used to prove identity. Some areas of DNA, called hypervariable minisatellites, have high variability between different individuals. If enough of these are compared, it can prove someone's identity beyond reasonable doubt. For example, if 20 of these areas are compared, then "less than one in 10^{12} people" will have no differences between their DNA for all of these areas (excluding identical twins). This can be used for paternity testing, and can also be used in criminal cases, either as evidence against the defendant or to eliminate someone from enquiries (Debenham 1990, pp.38-40).

2.2.2 Genomics

2.2.2.1 Background

2.2.2.1.1 The Genome

The genome is a term for all of the DNA that is stored in a cell's chromosomes (Primrose & Twyman 2004, p.21). The length of different organisms' genomes vary considerably, with the genome of a virus named Phi-X174 only 5386 bases long (DeSalle & Yudell 2004, p.40), and the human genome approximately 3 billion bases long, coding for between 20,000 and 25,000 genes (Jorde et al 2006, pp.3,7).

2.2.2.1.2 Sequencing the Genome

The Human Genome Project began in 1990, with the aim of sequencing and mapping the entire human genome (DeSalle & Yudell 2004, p.48). In 2003, the first complete sequence of a human genome was finished (Jorde et al 2006, p.3). The sequencing was achieved using a machine named ABI Prism 3700. This machine works on the same principles as the sequencing techniques described

in section 2.2.1, but with two major differences. Firstly, the bases were labelled with fluorescent colours (one for each base) rather than radioactivity. This allowed their order to be read by a computer using a laser, rather than by hand. Secondly, the fragments of DNA were passed through tiny, fluid-filled capillary tubes, rather than a gel, which greatly increased the amount of DNA that could be sequenced at a time. The ABI Prism 3700 can sequence up to 1,000,000 bases per day (DeSalle & Yudell 2004, pp.40,50).

2.2.2.1.3 The Future

The cost of sequencing a whole human genome has rapidly decreased since the completion of the Human Genome Project. At first, the cost, including money spent on developing the technology, was approximately three billion dollars (Robertson 2003). By 2007, this had reduced to approximately 10 million dollars, by 2008 to approximately one million dollars (Henderson 2009), at the start of 2010 to approximately 48 thousand dollars (Los Angeles Times 2010) and it is now available for as little as five thousand dollars (Cadwalladr 2013).[2]

In a paper published in 2009, Axelrod et al stated that it was likely that there would soon be a large increase in the number of genomes that had been sequenced. This is a prediction that has very rapidly come into fruition. When a review of the literature was first conducted in 2010, a comprehensive search found only nine online databases which contained a full human genome sequence, some of which were replicated between databases. Three years later, the 1000 genome project alone contains 1092 full human genome sequences (1000 genomes, 2013), and in 2012 it was announced that the NHS would aim to sequence the full genome of 100,000 patients within five years (Gov.uk 2012).

2.2.2.2 Benefits of Genomics

2.2.2.2.1 Amplification

One of the main benefits of genomics is that it amplifies the benefits of ordinary genetics. For example, genomics makes it easier for a disease gene, or the genetic factors that predispose people to complex disorders to be discovered (DeSalle & Yudell 2004, pp.119-123 and Primrose & Tywman 2004, pp.16-17). This is due to various reasons. One of these is the mapping of the location of thousands (and eventually all) genes on the chromosomes. This means that if the general location of a disease gene is known, the map can be used to find "candidate genes", which can then be tested; a much quicker technique than the pre-genomic practice of positional cloning (Primrose & Tywman

[2] As stated in footnote 1, since submission of this thesis a company named Illumina have released a machine that is claimed to have the ability to sequence an entire human genome for under one thousand dollars (Illumina 2014).

2004, p.16). One powerful new way of discovering which genes are involved in complex diseases is the genome-wide association study (Cornelis et al 2010). Genome-wide association studies involve analysing a very large number of genetic variants in a very large number of participants, to see if there is an association between any of the variants and a particular disease. Any variants associated with the disease may not themselves contribute to the disease, but they do identify the approximate location of disease-causing variants (Cowperthwaite et al 2010).

Screening is another benefit of genetics that could be amplified with genomics. In the future it may become feasible to scan someone's entire genome to assess their genetic susceptibilities. Unlike current screening, this could identify genetic susceptibilities to complex diseases as well as single-gene disorders. With complex diseases there are often preventative measures, such as increased exercise or changes to diet, that can reduce the risk of the disease, and so genomic screening may become an "effective medical tool" (DeSalle &Yudell 2004, p.129).

2.2.2.2.2 Other Benefits

Pharmacogenomics is the study of how variations in genomes affect interactions with medical drugs (NCBI 2004). Drugs have different levels of effectiveness between individuals, and cause side effects in some individuals but not others, because of variations in genes. Once it is understood which drugs are the most effective and safe for each genotype, genome sequencing will allow drugs to be tailored to the individual (DeSalle & Yudell 2004, pp.130-132; Primrose & Twyman 2004, pp.109-110). Research is currently underway in this area. For example, Ingle (2013) describes genome-wide association studies conducted to identify any genetic variations that affect the efficacy of endocrine therapy for women with oestrogen receptor-positive early breast cancer. They found several noteworthy associations that allowed them to investigate the effect that different alleles had on both treatment efficacy and side-effects. One early success of pharmacogenomics relates to the use of the anticoagulant drug Warfarin where alleles responsible for approximately half of the variation in the dose required in patients with European ancestry have been identified (Ritchie 2012). However, despite these findings clinical trials have so far failed to demonstrate any significant benefit of the use of genetic data in determining the administered dose when compared to standard clinical methods (Stergiopoulos and Brown 2014).

Environmental factors play a large part in complex diseases (DeSall & Yudell 2004, pp.132-133). For example, it has been known for a long while that smoking greatly increases the risk of lung cancer and many other diseases (Doll et al 2004). However, the extent to which harmful environmental factors affect individuals varies due to genetic variations. Eventually, genomics should be able to identify individual susceptibilities to environmental factors. It should also be possible to use

genomic information to better identify which aspects of the environment (such as chemicals) are harmful to humans, allowing people to be better protected from them and reducing the need for animal studies into toxicology (DeSalle & Yudell 2004, pp.133-135). Research in this area has already begun. For example, Chand et al (2014) conducted a study which suggests that genetic variations in pregnant women may alter the effect of organochlorine pesticide exposure on foetal growth and the risk that a foetus will be small for gestational age. Wang et al (2014) found that the risk of neural tube defects developing in a foetus due to exposure to indoor air pollution was affected by the mother's genotype for the CYP1B1 gene.

Not all of the benefits of genomics are medical. For example, genomics can be used to further the study and understanding of evolution, and can help with the eventual aim of creating a "tree of life" of all species on the planet (DeSalle & Yudell 2004, pp.95, 99-108). The genomes of a large number of species have now been sequenced, although this is so far only a tiny fraction of the total species in existence. For example, as of 2014, the genomes of 150 species of plants have been sequenced out of an estimated 435,000 total species of plants, the genomes of 235 terrestrial vertebrates and fish have been sequenced out of an estimated 80,500 and the genomes of only 98 insects have been sequenced out of an estimated 10 million (Azvolinsky 2014). Genomics can also be useful for helping to protect endangered wildlife. For example, it is illegal to sell wild specimens of the endangered St. Vincent Amazon parrot. Since genetic signatures are known for those birds which are bred in captivity, genomics can be used to determine whether a bird being sold is from one of these populations or from the wild (DeSalle & Yudell 2004, p.108).

2.2.3 Genetic Resources for the Public

There are many genetics resources available to the public; this section provides several examples.

Genetics Home Reference (found at http://ghr.nlm.nih.gov/) is a website that is run by the Lister Hill National Center for Biomedical Communications, which is itself run by the U.S. National Library of Medicine. It was created in 2003 in order to communicate genetic information (including information from the Human Genome Project) to the general public. It provides information about genetic conditions, genes, gene families and chromosomes, along with a handbook (which contains a wide variety of background genetic information, such as information on inheritance patterns and genetic testing) and glossaries. It also aims to help people to find information from external websites. Information is reviewed by external genetics experts before being posted (Mitchell et al 2006 and Genetics Home Reference 2010a). The website is easy to navigate, and supports both browsing and searching for information.

The Genetic Alliance (found at http://www.geneticalliance.org/) is an organization that advocates the use of genetics to improve health and who provide tools and information about genetics which are available to the public (Genetic Alliance [n.d.]a and Genetic Alliance [n.d.]b). For example, they provide a tool named Disease InfoSearch which allows the public to search for resources to do with specific genetic conditions. These resources include information about support groups and advocacy organisations, links to treatments, references and clinical descriptions and information from National Library of Medicine databases (Disease Infosearch [n.d.]). Another tool named 'Trust It or Trash It?' helps people to think critically about health information that they find (Trust it or Trash it? 2013). The Genetic Alliance also provide a "resource repository" to which the health community can upload genetics-related resources to become available to both the community and the public ((Resource Repository [n.d.].

The website for the National Human Genome Research Institute (found at http://www.genome.gov) provides a lot of genetic information aimed at the public. This information includes FAQs, factsheets about aspects of genetics and factsheets about different genetic diseases. It also provides a talking glossary and an online education kit.

The websites of providers of direct-to-consumer (DTC) genetic tests also provide information to the public; these are described in section 2.4.

There are also some websites that provide genetic information that is inaccurate. Many of these websites use this information to try to prove a point. For example, an article on a website named exchangedlife (2002) tries to use genetics to disprove evolution. Some of the genetics information in this article is accurate. However, one of the main arguments put forward is that humans needed to have gained 60 million "positive mutations" in "500-600 thousand years" to have evolved from apes, a rate of 10 per month: a very inaccurate statement.

2.3 General Legal and Ethical Issues Related to Genetics

There are many legal and ethical issues related to genetics and genomics; this section describes some of them.

2.3.1 Legal Issues

2.3.1.1 Genetic Discrimination

The Equality Act 2010 is the most recent anti-discrimination legislation in the UK. It had the aim of consolidating previous anti-discrimination legislation into a single piece of legislation, as well as

strengthening the law against discrimination. Genetic discrimination is not included in the act, and as such, genetic discrimination is still legal in the UK (Equality Challenge Unit 2010, Human Genetics Commision 2010a, Human Genetics Commission 2010b, BioNews 2006 and Equality Act 2010). With regard to genetic discrimination and insurance, the current legal position in the UK is that customers must inform insurers about any genetic susceptibilities that they have tested positively for. However, there is currently an agreed moratorium which means that customers do not have to inform insurers about genetic susceptibilities for policies "which fall within the financial limits of £500,000 for life insurance, £300,000 for critical illness insurance, and £30,000 pa for income protection policies". Also, in situations where the insurer does know of genetic susceptibilities they have agreed not to discriminate against individuals without justification (Wilkinson 2010, pp.283-284). The moratorium was originally due to expire in 2014 but has now been extended to 2017 (Wilkinson 2010; HM Government 2011).

In 2008 the USA passed the Genetic Information Nondiscrimination Act. This act makes it illegal for unions, insurers and employers to discriminate against individuals based on genetic susceptibilities highlighted by genetic tests. Employers are now not able to request or access the results of genetic tests, are not able to "hire, fire, promote or compensate an employee on the basis" of them, and health insurers now cannot "determine coverage, premium rates or increases/changes to terms based upon [them]". However, life insurance and long-term care insurance are not included in the act (BioNews 2008). Interestingly, the act was voted for unanimously in the senate, and passed by 414 to one in the House of Representatives (National Human Genome Research Institute 2010).

2.3.1.2 Genetic Patents

Patenting, in the area of biotechnology and pharmaceuticals, is generally considered necessary to encourage the sizable investment of time and expense often required for the development of new products and technologies in these fields. However, the practice of gene patenting is a controversial issue. Proponents of gene patenting argue that DNA is a chemical and as such any gene sequences that are identified with a function or disease association should be treated the same way as a newly identified chemical i.e. patentable for all known uses and any uses that come to be known in the future. Opponents argue, however, that DNA is created by nature and so can never be considered a human invention (Dutfield 2006). There are also concerns that the patenting of a gene can be used to stop any commercial research into the gene (Meek 2000) which can lead to monopolies of health products (Dutfield 2006). An example of this is provided by the US company Myriad Genetics who own patents related to the BRCA genes; certain alleles of which are well established as increasing the risk of breast cancer. Myriad genetics provides an expensive test to identify these mutations and

has attempted to use its patents to prevent any other company in the USA from providing alternative tests (Gold & Carbone 2010).

The legal position of gene patents differs depending on jurisdiction. Historically, gene patenting has been permissible in the USA (Dutfield 2006). However, a recent legal ruling by the US Supreme Court has altered that position. In a case brought against Myriad Genetics (whose BRCA patents are described above), the Supreme Court ruled in 2013 that unmodified genes are a 'product of nature' and hence cannot be patented (genome.gov 2014 and Supreme Court of the United States 2013). In the EU, gene patenting is permissible as long as the application contains a disclosure of the industrial use of the genetic sequence (European Patent Office 2013). A 2011 ruling by the UK Supreme Court confirmed this position and decreed that the disclosure of the sequence's industrial use need only be credible, not provable (Dehns 2011).

2.3.1.3 Data Protection Act

All organisations or individuals who "process personal data" have a legal duty to comply with the Data Protection Act 1998 (Information Commissioner's Office [n.d.]a). This means that certain principles must be followed. These principles include the requirement that "personal data shall be adequate, relevant and not excessive in relation to the purpose or purposes for which they are processed" and "personal data processed for any purpose or purposes shall not be kept for longer than is necessary for that purpose or those purposes" (Information Commissioner's Office [n.d.]b).

2.3.2 Ethical Issues

2.3.2.1 Privacy, Confidentiality and Informed Consent

For systems that hold information about individuals, respecting privacy and maintaining confidentiality are both important ethical principles, and are part of the Association for Computing Machinery (ACM) code of ethics. The code states that data should be accurate, with revision and correction possible for participants. All access to the system should first be authorized, and there should not be any disclosure of information to unauthorized individuals. The system should contain no unnecessary information, and information should only be used for the purpose for which it has been gathered. The length of time for which the information will be held should "be clearly defined and enforced" (Association for Computing Machinery 1992). An organisation holding information about someone should make sure that this information is protected and only accessible "by those with a need to know" (Reynolds 2006, p.126). An effective security system is essential to do this, but it should not involve too many trade-offs in usability (Schultz 2006, pp.111-113). Confidentiality

should be maintained, both when it has been explicitly promised and when it is implicitly required (Association for Computing Machinery 1992).

The Nuremberg Code is an ethical code for experiments involving humans. It was created after the Second World War, during the trial of Nazi doctors for experiments performed on concentration camp prisoners. It has "served as a blueprint for today's principles that ensure the rights of subjects in medical research". The code states that "the voluntary consent of the human subject is absolutely essential", and that this consent must be fully informed without any kind "of force, fraud, deceit, overreaching, or other ulterior form of constraint or coercion". Informed consent is now required by international law (Shuster 1997, p.1436).

The principles of privacy, confidentiality and informed consent are all important in genomics research. Privacy and confidentiality may even be more important in genomics than in other areas of research, as an individual's DNA cannot be changed (and so any loss of privacy or confidentiality is permanent) (Heeney et al 2010), and genome sequence information about one individual also reveals "probabilistic" genome sequence information about close family members (McGuire et al 2008, p.153). This probabilistic genome sequence information raises privacy, confidentiality and informed-consent issues about an individual's family. For example, Cassa et al (2008) demonstrate that using an individual's genotype and allele frequencies for the population, it is possible to estimate a sibling's whole genotype with a high level of accuracy (in some cases as high as 98.5%). It is also possible to discover an individual's identity by comparing their genome sequence to a relative's (McGuire et al 2008). This raises the question of whether or not relatives should be involved in the informed-consent process. McGuire et al (2008, p.154) argue that relatives should not need to give consent for the initial genome sequencing and research, but that any implications for them should be discussed with the participant, who should be encouraged to include them when making the decision about whether or not to give their consent for the research. They further argue that, with regard to consent for the release of genome sequence data, "investigators should take a family-centered approach" with any objections raised by relatives investigated and reviewed by ethics teams.

Heeney et al (2010, pp.1-2) argue that when genomic data from research is shared with other researchers, or is published with open access, simply "making the data non-identifiable" is not enough to guarantee that a participant will never be identified. This is because of the uniqueness of genomic information (that could allow, for example, a unique set of traits for a person to be identified), and the availability of large amounts of other data (such as socioeconomic or census) in "the data environment". If these sets of information are combined then a person's identity could be

discovered. The authors recommend that before any collections of genomic data are established, "privacy risk assessments" should be carried out in order to look at the whole "data environment" and how this could potentially be misused to identify participants. There are concerns that customers of providers of DTC genetic tests who are able (through the provider) to share their results with a social network may not be fully aware of the risk of loss of confidentiality (both individually and for their relatives as well) (Resnik 2009).

One of the major differences between genomics research and other clinical studies is that genomic information is usually stored in large databases designed for open-ended research in the future. This is compared to the (usually) specific hypotheses and specified study times of clinical studies. This could lead to potential breaches of privacy, either through the sharing of information at some point in the future, or through someone illegitimately accessing the database. One way to ensure that participants give fully-informed consent in respect to the sharing of information is to specify the required conditions for access to the database (e.g. only used "for approved research", only used if the data is properly protected etc.) (Roche 2009, p.295). McGuire et al (2008, p.155) argue that until there is a change in the mechanisms for informed consent with regard to this issue "genome researchers must ensure that research remains within the spirit of the original informed consent, or re-consenting should be considered".

Participants should be fully informed about the risks of genome sequencing before their genome is sequenced. These risks include discovering medical information about themselves and their families that they may not wish to know (including both non-preventable conditions and preventable conditions that may require action that they do not wish to take) and a potential loss of privacy (Robertson 2003).

2.3.2.2 Genomic Screening

One important ethical point raised by Robertson (2003, p.38) is that an individual's genome should only be sequenced if there are "good reasons to do so". This corresponds with a point made by Ransohoff and Khoury (2010), who argue that it would be a mistake to assume that having access to genomic information will always be beneficial. They argue that in situations where the identification of risk factors for disease is uncertain or inaccurate, or where it is accurate but there is no (or little) benefit from knowing it, then the knowledge may cause more harm than good. Robertson (2003) recommends that whole-genome sequencing should only be undertaken if it is shown to be more beneficial than testing small genome segments on an as-needed basis. He also argues that

mutations that would cause a disease that is currently untreatable should either remain untested or the patient should not be told about them.

Another issue is that people who find that they do not have any predispositions towards "common complex diseases ... may not appreciate that they still are at risk for the disease" (Glenn, 2007). Also, the incompleteness of current knowledge about the genetic causes of complex diseases must be taken into account. Although a current screening test may give a negative result for a certain disease, there is no guarantee that this is the whole picture; future discoveries of mutations associated with the disease may mean that someone who thought that they had a low risk for it may actually still have a high risk (The Lancet Oncology 2008).

2.3.2.3 Children and Families

Wilfond and Ross (2009) argue that the decision on whether a child's genome should be tested (i.e. to identify genetic disorders) should be left to the parents. They argue that most medical decisions about a child's health are currently taken by the parents, and that since genomic testing has the potential to provide not only medically useful information, but also information that is not medically useful (or may be useful in later life, but not during childhood) and may cause anxiety or change parenting behaviour, the issue is too complex for mandatory or restrictive policies. Robertson (2003, p.40) thinks differently, stating that children should only have their genome sequenced if "there is a strong medical justification for doing so", regardless of any parental desires for sequencing. He adds that if there is a need for genetic testing, smaller-scale tests should be used if adequate. Also, if a child's genome is sequenced for a medical reason, the information should be "erased or stringently protected" after it has been used. Robertson believes that these principles should also apply to foetuses and people "who lack present competency to consent".

If an individual whose genome has been sequenced has been found to have a substantially high relative risk for a disease, but he/she does not wish to inform their families, an ethical problem exists over whether family members who are likely to be at risk themselves (due to shared genetics) should be informed anyway. For example, Loud et al (2006) describe a case where a woman and her siblings undertook genetic testing due to the discovery of a mutation in her father's genome that substantially increases the risk of breast cancer. Although she tested positive for the mutation, she did not want to inform her family, and lied to them about the result. This meant that her children thought that they had no chance of inheriting the mutation, when in reality they had a 50% chance. Since her eldest daughter was nearing the age at which breast cancer screening would be recommended for individuals with the mutation, the research team was faced with a difficult ethical

decision. After an ethics consultation it was decided that the mother's right to confidentiality was greater than her children's right to be informed of an increased risk of disease. However, the research team felt that they should encourage her to tell her children (Loud et al 2006). Offit et al (2004, p.1469) argue similarly, stating that "health care professionals have a responsibility to encourage but not to coerce the sharing of genetic information in families". However, it may be ethical to inform relatives if "the harm [of not informing them] is serious, imminent and likely; prevention or treatment is available; and ... [if] a health care professional in like circumstances would disclose" (Knoppers et al 1998, p.484).

2.3.2.4 Other Ethical Issues

As genomic knowledge increases, there is a danger that people may be discriminated against (e.g. when purchasing health insurance) based on aspects of their genome (such as genetic susceptibilities) (Heeney et al 2010). Although there are some specific cases in employment where an individual's genome makes them extremely susceptible to being injured in a specific industry, and so genetic testing is ethical, it is generally felt that it is unethical to discriminate against someone due to their genetics. Also, the possibility of genetic discrimination may discourage people from having their genomes sequenced in the first place (Robertson 2003).

With regard to law enforcement, the use of forensic DNA techniques and genetic databases to generate groups of potential suspects for further investigation can lead to "false positives" (Heeney et al 2010, p.6). This would obviously be an unethical use of genome-sequence information. In a similar vein, Robertson (2003, p.40) states that information that has been obtained by sequencing an individual's genome "for medical purposes" should not be used for law enforcement purposes.

2.4 Direct-to-Consumer Genetic Tests

2.4.1 Background

2.4.1.1 Introduction to Direct-to-Consumer Genetic Tests

Sections 2.2.1.6 and 2.2.2.2 described both the current and potential future benefits of genetic testing with regard to health. These benefits, combined with the advances in genomic sequencing described in section 2.2.2.1.2, have led to the rise of direct-to-consumer (DTC) genetic testing.

DTC genetic tests are genetic tests that are commissioned directly from private companies by consumers, normally with no need for the involvement of health care professionals. Consumers are instructed to post a sample (usually saliva) to the company, which sequences the DNA, analyses the

code and returns a report (Bloss et al. 2011a; Genetics Home Reference 2010). Several different types of DTC genetic tests are available, including pharmacogenomic, nutrigenomic and ancestral. However, this section, and the thesis as a whole, is focused on DTC genetic tests that provide relative genetic risk assessments for multiple diseases; these shall simply be referred to as DTC genetic tests.

Unlike the full genome sequencing mentioned in section 2.2.2.1.2, providers of DTC genetic tests sequence only part of the genome; providers will sequence either a large number of locations in the genetic code that are known sites for potential mutations, normally SNPs, or focus on certain specific disease mutations. Any identified mutations that are associated with disease are then used to estimate the consumer's relative risk of suffering from a large number of diseases (Wasson 2009; Griesmann 2009; Samuel et al 2010; Korf & Irons 2013, pp.214-215).

Providers of DTC genetic tests claim that they have certain benefits for the consumer. For example, 23andMe (one of the biggest DTC providers) claims that providing relative risks for diseases will allow participants to prepare for and watch out for diseases that they are at a high risk of contracting, as well as enabling them to "make better lifestyle choices". They state that their test will also allow people to know if they are carriers of certain diseases (such as sickle cell anaemia) and how their genetics may affect certain medications (23andMe 2013a). They claim that the test will inspire participants "to take more responsibility for their own health and well-being" (23andMe 2013b). Inherent Health (another provider of DTC genetic tests) claims that their test will "empower consumers" and help them to prevent age-related chronic diseases (Inherent Health 2009).

2.4.1.2 Criticisms and Concerns

Despite the beneficial claims made by providers, there are many concerns about, and criticisms of, DTC genetic tests in the literature. For example, one concern is that providers' assessments of disease risk are often not based on properly-validated links between mutations and disease, and hence may be inaccurate (Murray et al 2010). Since many providers do not publish the false result rate it can be impossible for health professionals to determine their reliability (Wasson et al 2006). It is argued that any inaccuracies would be profoundly unethical as false positives can cause anxiety and lead to "unnecessary preventative measures", and false negatives may encourage unhealthy behaviour in people who may in fact need to be especially careful (Samuel et al 2010, pp.221-222). This potential for inaccuracy was highlighted by a reporter for the Sunday Times, who commissioned a test from three different companies and received differing and often contradictory risk assessments (Fleming 2008). Also, many of the tests are "based upon very modest gene-disease

associations"; even if they accurately show a statistically significant increase in a person's risk for a complex disease, it may not be large enough to be clinically significant (Samuel et al 2010, p.221). Even if the tests are accurate, an individual's actual risk of developing a disease (when factors such as family history and environment are taken into account) could be very different from their relative risk based on their genetics. This may lead to "a false sense of security" in people who do not have a high relative risk but actually do have a high actual risk (Kaye 2008, p.2). Ameer and Krivoy (2009, p.887) state that "currently ... there is limited ability to predict the risk of diseases based on genetic profiles and genomic expression patterns".

Another concern is that, even if the tests are accurate, it is uncertain how or if the information produced will affect behaviour. Although it is "intuitively appealing" that knowledge of genetic risk information would modify behaviour, "evidence to support it ... is scanty" (Hunter et al 2008, p.106) and it is currently uncertain "when or how genetic risk information might motivate healthy behaviour" (Henrikson et al 2009, p.2). Indeed, arguments can be proposed for why this information may have no effect. For example, if an individual has only a slightly increased risk of a disease then "aggressive prevention measures" would be unethical. However, healthy behaviours reduce the risk of a large number of diseases for everyone, regardless of their genetic risk (Henrikson et al 2009, p.2). Since the majority of mutations tested only have a small effect on relative risk (Hall & Gartner 2009) it is far from certain that a small increase in relative risk for one disease will cause an individual to pursue healthy behaviours that a health-conscious individual should already have been pursuing.

The information provided by the websites of the companies that sell DTC genetic tests has also been criticised (Borry et al 2013). This is an important criticism for two reasons. Firstly, in order for customers to fully consent to any research involving their DNA, they need to be informed of and understand a range of information, such as the testing process and impact of results (Wasson 2009). Secondly, DTC genetic tests often bypass health care professionals. Evans (2009, p.172) argues that "without the intermediary of a health care provider to validate the analysis and contextualize the risk, these tests can have an alarming and bewildering effect on consumers". Also, other factors such as diet and family history need to be factored in to properly interpret results (Lee & Crawley 2009), and people may expect health care services to provide interpretive help, thus increasing health care costs (Caulfield 2009). Therefore it is important for the companies to provide as much information as they can to help minimise the risk of confusion and misinterpretation, to ensure that consumers are fully informed when giving consent and to prevent a large increase in the use of health resources.

Other concerns focus on ethical issues. For example, if DTC genetic tests do confer health benefits, then it may increase health inequality, as both access to the internet and enough money are required to purchase one (Wasson et al 2006). It is impossible for the provider to guarantee that the individual commissioning the test is sending in his/her own DNA sample; therefore it is perfectly possible for someone to send in another person's DNA without his/her knowledge, and thus obtain illegitimate access to very personal information (Samuel et al 2010). As with all genetic testing there is a danger that people may be discriminated against (e.g. when purchasing health insurance) based on aspects of their genome (such as genetic susceptibilities) if their results were to become known (Heeney et al 2010) and that close family members' genetic information could be deduced from an individual's results (McGuire et al 2008).

2.4.1.3 Regulation of Direct-to-Consumer Genetic Tests

Regulation of DTC genetic tests is a contentious issue, and many professional organisations and researchers have suggested regulations that they believe should be enacted. For example, the European Society of Human Genetics has published an exhaustive list of suggestions, such as the requirement that tests should be clinically useful, that laboratories "should comply with accepted quality standards" and that relevant information should be provided to the consumer before any testing. (European Society of Human Genetics 2010, p.1271). In contrast, Wright et al (2011, p.295) believe that only five regulatory areas are needed. These would be in relation to ensuring that the consumer has fully consented to the test, that the laboratory is fully accredited, that any associations between genes and diseases are backed up by evidence, that the results are interpreted by staff with appropriate qualifications and that the companies are prevented from making "false or misleading claims".

In terms of actual regulation, the legal position differs from country to country. Several provisions of European Union (EU) law are relevant to DTC genetic tests. These mainly relate to data protection, anti-discrimination issues and consumer-protection issues (Soini 2012; Borry et al 2012). There is no "specific genetic legislation at EU level" (Soini 2012, p.145), however, the proposed 'Regulation on In Vitro Medical Devices' directive may alter this situation, with amendments put forward by one MEP that would require genetic counselling to be provided with every genetic test (ESHG 2014).

Within the EU, there is variation in how national laws deal with DTC genetic tests. For example, genetic tests are banned in France apart from for certain reasons (such as medical) within which informational purposes are not included. Permitted genetic tests can only be performed with the consent of a healthcare professional with whom an individual has a "medical relationship", and

individuals are actively prohibited from ordering a genetic test that does not conform to the requirements of the law. In Germany genetic tests can only be performed by a doctor, and individuals must give informed consent after presentation of sufficient information. Although the first requirement clearly prevents DTC testing companies from operating within Germany, individuals are not prohibited from ordering tests from other countries. The situation in the UK is substantially different to France and Germany. The only relevant UK law that relates specifically to genetic testing is a ban on the genetic analysis of a sample without the individual's consent. Other than this, DTC genetic tests are only subject to general laws on the sale of products and medical products e.g. consumer protection laws (Borry et al 2012, p.716).

In the USA the regulatory situation is far from decided. There is currently no federal regulation of DTC genetic tests, which are permitted in a majority of states but prohibited in a minority (Tamir 2010). In 2010 the Food and Drug Administration (FDA) warned companies "that it felt such tests needed regulation"; however, this regulation is yet to be codified. In 2012, 23andMe became the first provider of DTC genetic tests to file for FDA clearance for a small number of its genetic tests (Allison 2012, p.1027)[3].

2.4.2 Research

Research into DTC genetic tests has until relatively recently been fairly thin on the ground. However, in the previous two to three years there have been a number of studies. All identified research into DTC genetic tests is presented in this section.

2.4.2.1 Effects of and Responses to DTC Genetic Tests

One of the most significant areas of research into DTC genetic tests has been research into participants' responses to the tests, and the effect that the results have had on them. One early study was conducted by Gordon et al (2012) who interviewed 60 participants who had been provided with a free test that analysed genetic risk for eight different diseases. Approximately one third of their participants claimed to have made behavioural changes, including an increase in exercise, an increased diligence in the use of sunscreen and bringing high risk areas of their results to the attention of a healthcare professional. Similar results were found by Kaufman et al (2012) who conducted a large-scale survey of 1048 actual consumers of DTC genetic tests, contacted through three of the main providers (Navigenics, deCODEme and 23andMe). One third of participants claimed that after receiving their results they were taking more care with their diet, 14% claimed that they had increased their level of exercise and 16% claimed to have made changes to medication

[3] See 9.5 Postscript: 23andMe

or supplements. In contrast to these two studies, Bloss et al (2010; 2011b; 2013) found no significant effects of the tests on participants. Similar to Gordon et al (2012), participants in Bloss et al's study were not actual consumers; their study population consisted of 2037 participants who had been invited to purchase a test at a heavily-subsidized rate and given free access to genetic counselling. Comparisons with baseline levels three months and a year after receipt of results found no significant changes in participants' level of anxiety, exercise behaviour or the amount of fat consumed in their diet. The discrepancy between the results of the latter study and the first two are interesting. The difference in the results may be due to a difference in the study populations. Alternatively, changes to health behaviour may not have been identified in Bloss et al's study due to the narrow focus; only two health behaviours were measured compared to the open-ended questions used by Gordon et al (2012) and Kaufman et al (2012).

As described further in section 2.4.2.3, Wasson et al (2013) undertook a small-scale study in which 20 participants were provided with a free DTC genetic test and interviewed four times over the course of a year. Most of the results related to participants' opinions about the tests; however, some results did focus on participants' psychological and behavioural reactions. Psychological reactions were varied soon after receiving results but after three months the majority stated that their experience had been positive. After a year, half stated that they had had no psychological reaction to the results and several stated that they had had a positive reaction. Several participants reported that they had made positive changes to their health behaviour; most commonly to do with exercise or diet. One of the main reasons given for changes to health behaviour was maintaining the 'low-risk' found in their results.

In contrast to the generally positive or neutral effects reported in the studies mentioned above, Mahon (2012, p.260) presents three case studies of women who had discovered a highly-increased risk for breast cancer and ovarian cancer in their DTC genetic test results. She describes how their results caused them "psychosocial distress" and that they subsequently needed counselling support, a service not provided by their test providers. Results from case studies should always be treated carefully; however, it is interesting to note that two of the individuals in her report had been purchased a DTC genetic test for a present, and had therefore possibly not given fully-informed consent.

Researchers at 23andme assessed the effects of discovering the presence of one particular type of mutation in consumers' results; BRCA mutations that greatly increase an individual's risk of breast and ovarian cancer. Studies by providers should always be read with caution due to the inherent conflict of interest. Also, this study examined the effect of the discovery of one mutation that is well

established as causing a large increase in relative risk; this is a substantially different situation from the small changes to the risk of a large number of diseases that DTC genetic tests normally show. Interviews were conducted with (amongst others) 25 individuals who had learnt from their results that they carried one of these mutations. No participants suffered from extreme anxiety, and 11 had a neutral response to the findings. However, four participants did report transitory moderate anxiety. Many positive actions were carried out after receipt of results, including seeking medical advice and informing relatives of risk. Importantly, no participant who had been found not to have the mutation reported the cessation of any healthy behaviour or action (Francke 2013).

Several studies examined effects of the tests not solely related to health behaviour and anxiety. Vernez et al (2013) studied 10 students who had taken a test as part of a genetics course. The participants reported a positive motivational effect with regard to learning the course, and an increased engagement in areas such as ethics, benefits and risks and social and policy issues. Although participants reported intentions to make small changes to health behaviour, only one student followed through with them in a significant way. In a study by Kaphingst et al (2012, p.681), 199 participants were given a free genetic test that covered eight diseases. They found that over 80% of the participants could correctly recall their results. Participants were also "unlikely to interpret genetic results as deterministic of health outcomes", with those possessing the least deterministic attitudes significantly more likely to be white, have a higher level of education and to have found the results less confusing. James et al (2011) conducted a study in which patients of a preventative medicine clinic were offered a free DTC genetic test. Participants were randomly assigned to either a control group, who received their usual care, or a test group, who received their usual care with the addition of a test. When asked to rate their risk of contracting various conditions, those who received the tests initially gave a significantly higher rating of their risk for four conditions and a significantly lower rating for one, with no significant difference for 12 others; however, all of the significant differences disappeared on reassessment a year after testing.

For completeness, a pilot study by Bansback et al (2012) should be mentioned. However, little useful information can be gleaned from the results due to its unusual methodology. Three hundred and nineteen participants were presented with a hypothetical set of test results and asked to predict what their response would have been had they been actual results. Since participants were not given results that had anything to do with their own health, it is difficult to see how the findings are relevant to understanding how individuals who have received actual results would react. Nevertheless, the study found that 63% of participants thought that they would arrange an appointment with a doctor, 57% thought that they would make changes to their lifestyle, 57%

thought that they would undergo a screening test and 40% thought that they would have an increase in health anxiety.

Finally, the research in this thesis has contributed to the literature in this area in two publications: a conference presentation (Egglestone et al 2012) and a journal paper (Egglestone et al 2013).

2.4.2.2 Informational Aspects of DTC Genetic Tests

Another significant area of research has been research into the informational aspects of the tests.

Several content analyses of the information provided on the websites of providers of DTC genetic tests have been conducted. One early study was undertaken by Geransar and Einsiedel (2008, p.13), who analysed the websites of 24 providers in order to assess their "information provision and access requirements". They found that providers of tests with a medical focus were more likely to require clinician mediation than providers of enhancement testing. The former were also more likely to recommend that counselling be arranged by a doctor, whereas the latter were more likely to recommend long-distance counselling. In another study, Einsieldel and Geransar (2009, p.354) analysed the advertising information of a large number of providers. They found that information about who could be considered at risk for a disease and the factors that cause disease to be "limited, vague or inaccurate". The websites overemphasized the benefits of taking the test and the risks of not taking them, whilst not providing enough information about the risks of taking them. Similar negative findings can be found in other content analyses. Lachance et al (2010, p.310) assessed both the information provided, and the usability of that information. They found a wide variation in the amount and usability of information provided, and stated that it is "apparent that most users would struggle to find and understand the important information on most sites". Hennen et al (2010, pp.180-181) found that a majority of websites did not "meet a minimum set of quality criteria" for the information they should provide, leading to "fundamental information deficits". Singleton et al (2012, p.433) analysed the number of statements on benefits, risks and limitations on the websites of 23 providers. They found an average of six times as many benefits statements as statements of risks and limitations. The benefits that were most frequently stated were "disease prevention.... consumer education....personalized medical recommendations and....the ability to make health decisions". Although 78% of websites did state a limitation, a risk was only stated by 35%. Liu and Pearson (2008, pp.135, 138-139) found that a large minority of DTC company websites did not provide information (or links to information) about basic genetics, a majority did not provide information about the "probabilistic nature of genetics" and a large majority did not provide information about "potential harmful effects of genetic testing".

The quality of information provided on company websites is obviously important for consumers' informed consent and understanding of the tests. Aside from these, it may also influence consumers' opinions of the tests and their decision to purchase. Sweeny and Legg (2011, p.1259) performed an experiment in which 99 participants were given information to read about DTC genetic tests that was either positive, negative or both. There was no significant difference in intent to test between those who read only negative information and those who read both positive and negative. However, there was a significant association between intent to test and the information that participants were given to read, participants' perceptions of the barriers to, and benefits of, taking a test and participants' "anticipated regret". A similar study is described by Kaphingst et al (2010, p.41), wherein 526 participants were offered, and asked to decide if they wanted, a free genomic test after visiting a website that provided all of the information it was considered they needed to know to make an informed decision. They found that, on average, those participants who had viewed more pages on the website found the decision easier to make than those who had viewed fewer pages. Also, those who found the decision easy to make "perceived the website information more positively overall than those who rated their decision as difficult".

Even if suitable information is provided it may not be easily remembered. In the study by Vernez et al (2013) mentioned above (section 2.4.2.1) where 10 students were provided with a DTC genetic test as part of their course, no participant was able to accurately remember the consent form details on the provider's website. Also, although eight participants agreed to 'biobanking', no participant could remember the length of storage time for the samples, whether or not personal data would be kept along with the sample or whether samples could be withdrawn.

In a study by Paquin et al (2012), 1959 participants were given a free genetic test for eight common diseases. A website with information about genetics and genetic testing was made available as part of the study; participants' likelihood of using it after receipt of test results was assessed. Participants were also assessed for different personality traits; the results showed that there was a correlation between conscientiousness and use of the website and, after controlling for demographics and perception of test results, there was a negative correlation between openness and use of the website.

Ducournau et al (2011, p.95) studied the marketing of DTC genetic tests. After analysing providers' websites they identified three areas that the marketing focused on. The first of these areas was "healthism". This is defined as the idea of health and hygiene being at "the top of the social values". The second marketing area was a focus on individuals as "actors of health decisions". The final area was identified as "the need for bio-social relationships". In a similar study, Arribas-Ayllon et al

(2011, p.53) analysed the websites of three of the main providers (Navaigenics, 23andMe and deCODEme) and identified "three distinctive registers" of personalisation in the websites. These were "a paternalistic (medical) register; a traditional (scientific) register and a democratic (consumerist) register".

Section 2.3.2.3 describes the ethical concern about genomic testing of children. Borry et al (2010, pp.52, 54) analysed the websites of 29 DTC genetic testing companies in order to assess their policies towards this. They found that 13 websites did not have any information about the testing of children, eight companies allowed parents to test their children, four websites stated that they were "not directed to children under 18 years" and four websites suggested that consumers "should have reached the age of legal majority" before testing. In a similar study, Howard et al (2011, p.1122) conducted a survey of 37 providers in order to assess their policies on the testing of children. They only received responses from 13 providers and concluded that "a clear majority of companies do perform genetic testing in minors".

Aside from the websites of providers, consumers may also discover information about DTC genetic tests in the media. Lynch et al (2011, p.486) analysed the media coverage of DTC genetic testing in the USA between 2006 and 2009. They found 92 news stories on the tests. In general these stories contained a moderate level of determinism (i.e. that genetics are deterministic in health outcomes) but "were neutral about validity and utility". Insurers and employers were indicated as the most likely cause of discrimination and providers and doctors as the most likely groups to violate an individual's privacy. After the passage of the American Genetic Information Nondiscrimination Act (described in section 2.3.1.1) most stories did not mention the law, although many stories "claimed lack of regulation would harm consumers".

2.4.2.3 Awareness and Opinions

One of the largest areas of research about DTC genetic tests has been into the awareness of the public about the tests and the opinions of the public, participants and consumers about them.

A study by Finney Rutten et al (2012) compared the US public's awareness of DTC genetic tests between 2008 and 2011. Their findings indicated a significant increase in awareness between these two dates, from 29% to 37% of the study sample (7674 in 2008 and 3959 in 2011). This difference remained significant when sociodemographic variables were taken into account. Those with a significantly higher level of awareness included individuals aged between 50 and 64, individuals aged between 65 and 74, university graduates, individuals who had access to health care, individuals who had previously been diagnosed with cancer, individuals who are 'online' and individuals who live in

an urban area. A similar study by Kolor et al (2012, p.860) found that there had been an increase in awareness of DTC genetic tests in the USA between 2006 and 2008. Their study compared the awareness of individuals from different states with a combined sample size of 16439 respondents; they found that Oregon was the most aware state with 29.1% awareness, and Michigan the least aware state with 15.8%. Their results showed an association between awareness of DTC genetic tests and "higher education, higher income and increasing age, except among those 75 years or older". The number of respondents who had used a test was smaller than 1%; approximately half to three-quarters of respondents who had used a test had shared their results with a healthcare professional. Goddard et al (2009) report on a similar study in 2006 that assessed awareness in three states with a combined sample size of 9807 respondents. They also found that Oregon was the most aware, with 24.4% awareness, and Michigan the least aware with 7.6%. When analysing predictor variables they found that a higher level of education, increased income and higher age (apart from for individuals aged 65 or older) were all predictors. In 2009, Ortiz et al (2011) assessed the awareness of DTC genetic tests amongst adults in Puerto Rico. They found that out of 611 respondents, 56% knew about the tests and four percent had taken one. Awareness was negatively associated with not having married and smoking, and positively associated with having looked for information about cancer in the past. Howard and Borry (2013) investigated European clinical geneticists' awareness and opinions of DTC genetic tests. They found that out of 131 respondents, 86% knew about the tests and that more than a third had had a patient ask about them. Although most disapproved of the tests, over 85% would offer counselling to consumers.

Two studies compared the awareness of different groups. Hall et al (2012) conducted a survey to determine if individuals with a high risk of cancer (i.e. individuals who had personally experienced cancer or had a relative who had done so) had a higher awareness of DTC genetic tests. They found that out of 1267 respondents, 49% of high-risk individuals were aware of DTC genetic tests, which was higher than the control group or the results of previous population-based surveys. However, the level of interest in testing was similar, although it was higher amongst those (and the relatives of those) who had self-referred to a cancer registry. Langford et al (2012, p.440) compared the awareness of DTC genetic tests among 6754 Hispanic, Black and White respondents. They found no significant difference between White and Hispanic respondents, but that Black respondents were significantly less aware than White respondents when sociodemographic variables were controlled for; although this significance disappeared when adjusted for numeracy. When respondents were taken as a whole awareness was significantly correlated with a higher level education, a higher income, increased age and various "numeracy variables".

An early study about individuals' opinions of DTC genetic tests by McGuire et al (2009, p.3) consisted of a survey of 1087 users of social media. Six percent of participants reported that they had used a DTC genetic test. In total, 74% stated that they would use a test "to gain knowledge about disease in their family" and 34% would consider it to be akin to a medical diagnosis. Seventy-eight percent of those who would be interested in testing would want their doctor to help provide interpretive help. In another study of individuals' opinions, Rahm et al (2012, p.448) conducted ten focus groups with "members of a large managed care organization". Their findings indicated that participants generally had a negative opinion of the tests but thought that some aspects would be useful. The two main areas of responses in the focus groups were the prevention of disease and the uncertainty about how consumers would react to results. In the study of the effects of the tests on 10 students mentioned above (section 2.4.2.1) conducted by Vernez et al (2013), participants were asked their opinions about the tests. The majority thought that they did not have much clinical use in the realm of disease risk and traits, but that pharmacogenic (genetic influence of drug response) results would be more useful. All of the students stated that in the same situation they would choose to do the genetic testing again. In an early paper on the study conducted by Bloss et al mentioned above (section 2.4.2.1) Bloss et al (2010, p.556) report on participants' responses when offered a subsidised genome test. In total, out of 2037 participants, 49.7% had concerns about being tested, with women, those who worked for health care organizations, younger participants, people with a higher level of education and people with "higher trait anxiety" more likely to have them. Gollust et al (2012, p.22) assessed the opinions of 369 participants who had enrolled for the Coriell Personalized Medicine Collaborative (a DTC genetic testing study). They found that, in general, participants decided to participate due to curiosity and a desire for the test to help improve their health. Fewer than 10% of participants thought that genetic risk was deterministic, although 32% "had misperceptions about the research study or personal genomic testing". The majority thought that the study would have health benefits, and the vast majority (92%) planned to share results with their doctor. In the same survey as that conducted by Kaufman et al mentioned above (section 2.4.2.1) Bollinger et al (2013) investigated consumers' opinions of DTC genetic tests. However, in this instance they focused on the regulation of the tests. They found that two thirds of the 1046 consumers surveyed thought that the tests should not be subject to oversight by the government. However, the vast majority thought that the scientific validity of the companies' claims should be monitored by an agency; 84% were of the opinion that a nongovernmental agency would be suitable and 73% that a governmental one would be. Almost all of the consumers thought that there should not be access to their information for insurance or law-enforcement purposes. Almeling and Gadarian (2013) conducted a survey of 2100 American adults to investigate opinions about genomics

issues. They found that 65% thought that DTC genetic tests should be explained with the help of health professionals. In a slightly different type of study, Su et al (2011, p.135) performed a qualitative analysis of 120 individual consumers' stories on blogs and DTC websites in order to discover their motivations and expectations; the five main areas found were "health...curiosity and fascination...genealogy....contributing to research and ....recreation".

Wasson et al (2013) conducted a study to assess participants' opinions and reactions to DTC genetic tests. As described in section 2.4.2.1, they interviewed 20 participants who they provided with a free test and genetic counselling. They found that a minority of consumers had small concerns about the tests, such as privacy issues or worries about receiving bad results. Three main reasons for taking the test were identified: curiosity, receiving health information that could be acted upon and altruistically helping either relatives or research. Roughly half of the participants mentioned that they were in some way uncertain about what to expect and roughly half that they had prepared themselves for what to do if their results were bad. Most participants understood the results; roughly equal numbers stated either that they would have been able to understand the results eventually without genetic counselling or that genetic counselling was essential to their understanding.

Leighton et al (2012) compared the interpretation of genetic test results by 145 members of the general public and 171 genetic counsellors; a survey was posted on Facebook with four example test results and relevant questions. There was a significant difference between the two groups' interpretations, with genetic counsellors considering the results as less helpful than did the general public. Although the general public mainly thought that the results were easy to understand, they often made mistakes in their interpretation.

2.4.2.4 Counselling and Healthcare Professionals

A final area of research into DTC genetic tests is research to do with counselling and healthcare professionals.

Several studies have examined healthcare professionals' opinions of DTC genetic tests. Brett et al (2012) surveyed 168 clinical geneticists and genetics counsellors in Australasia. In total, only 7% of respondents were confident that they would be able to accurately interpret test results and explain them to patients. Only 11% of respondents had been referred a patient who had purchased a DTC genetic test. In their experience, nearly all patients (92%) did not question the validity of their results, but most (80%) did require interpretive help. In a similar study, Hock et al (2011, p.325) surveyed 312 genetic counsellors. Most respondents (83%) had two or fewer enquiries about DTC

genetic tests, and only 14% had been asked for help in interpretation or discussion of the results. Just over half of respondents (55%) thought that genetic counsellors are obligated to have a knowledge of DTC genetic tests, and just under half (48%) thought that they should interpret results for patients. Approximately half (51%) thought that genetic testing should only occur in a clinical setting, although 56% thought that DTC testing was "acceptable if genetic counselling is provided". Mainous Lii et al (2013) assessed family doctors', rather than genetic counsellors', opinions of DTC genetic tests. Over half (58.1%) of the 1404 doctors who responded thought that DTC genetic tests would be more likely to harm than help patients' decisions about their health, although 31.6% thought that they would probably make no difference. Over two thirds (70.5%) of respondents had never been asked about DTC genetic tests by patients; 27.9% had only been asked rarely. In a similar study, Ram et al (2012, p.14) surveyed GPs in New Zealand. Just under half (47.8%) of the 113 GPs who responded had heard of DTC genetic tests. The biggest benefit of the tests was thought to be convenience; the biggest risks were thought to be "misunderstanding of results and inadequate provision of information". Respondents thought that the most appropriate individuals to provide genetic counselling would be genetic specialists, and that GPs themselves may struggle to do so given constraints on time, knowledge and experience. Respondents were also generally of the opinion that there should be regulation on the advertising of DTC genetic tests similar to that employed for prescription medicines. Powell et al (2012) assessed the self-reported educational needs of primary-care physicians in North Carolina with regard to DTC genetic tests. Only 39% of the 382 physicians who responded were actually aware of DTC genetic tests. A large majority (85%) did not feel that they were prepared to answer patients' questions about the tests and 74% wanted to learn more. Out of those who were aware of the tests, less than half (43%) thought that they were clinically useful.

Two studies attempted to assess consumers' actual use of healthcare professionals. The first, Giovanni et al (2010), surveyed 133 healthcare professionals. In total, 22 patients were referred to one of the respondents: 13 by self-referral, seven by another healthcare professional, one by an insurer and one by the provider of the test. Over half of respondents to whom a patient had been referred described the test as useful. However, single gene tests for BRAC mutations were included in this number which may have influenced the results; 85.7% of respondents thought that BRAC testing was useful, whereas 64.3% thought that the other tests were not useful. Respondents were asked to quantify the downstream costs of their consultations with patients; costs varied from $40 to $20,604. The second study, Darst et al (2013, p.335), assessed the use of genetic counselling by participants in the Scripps Genomic Health Initiative study, who had purchased a subsidised DTC genetic test from a provider named Navigenics, who had provided free genetic counselling. In total,

only 14.1% of the 1325 participants who responded had utilised the genetic counselling service. However, the percentage varied significantly between times when Navigenics ran a counselling-outreach programme and when they did not; 12.1% of participants utilised the counselling service when there was no outreach programme compared to 27.3% who used it when there was. Over half of participants who utilised the counselling service reported that it "improved their understanding of their results". The small usage of the counselling service in this study is echoed in the study by Vernez et al (2013) described above (section 2.4.2.1) where ten students were given a DTC genetic test as part of their course; only one student out of the 10 was interested in the free genetic counselling provided.

In a slightly different study to those above, Harris et al (2013, p.277) examined the websites of 20 providers of DTC genetic tests to examine their representation of genetic counselling and the expertise of genetic counsellors. They found that the picture of genetic counsellors portrayed on the websites were different to the traditional roles of genetic counsellors; genetic counsellors were portrayed to have the roles of "genetics educator; mediator; lifestyle advisor; risk interpreter; and entrepreneur".

Lovett et al (2012) analysed the websites of 20 providers of DTC genetic tests and found that only one company clearly offered genetic counselling. They also assessed the suitability of the tests for screening purposes based on evidence-based guidelines, finding that only four out of the 127 tests were suitable for use for general screening and only 19 were suitable for use for screening for specific groups.

2.5 Summary

The study of genetics has led to many benefits, from population screening (Jorde et al 2006, p.278) to the beginnings of gene therapy (Jorde et al 2006, p.296). The field has rapidly advanced from the discovery of the structure of DNA in 1953 (DeSalle & Yudell 2004, pp.21-22) to the first DNA-sequencing technique in the 1970s (DeSalle & Yudell 2004, p.40) and the complete sequencing of the Human Genome in 2003 (Jorde et al 2006, p.3). The new field of genomics that has arisen from this achievement promises to provide still greater benefits. It has already made it easier for a disease gene, or the genetic factors that predispose people to complex disorders, to be discovered (DeSalle & Yudell 2004, pp.119-123; Primrose & Tywman 2004, pp.16-17), and in the future is likely to allow drugs to be tailored to the patient (DeSalle & Yudell 2004, pp.130-132; Primrose & Twyman 2004, pp.109-110) and make it feasible to scan someone's entire genome to assess their genetic susceptibilities to both complex diseases and single-gene disorders (DeSalle &Yudell 2004, p.129).

However, there are many legal and ethical issues that must be considered, such as privacy, confidentiality and discrimination (Heeney et al 2010).

The potential benefits of genomics have led to the rise of DTC genetic tests; genetic tests that are commissioned directly from private companies by consumers, normally with no need for the involvement of health-care professionals (Bloss et al. 2011a; Genetics Home Reference 2010). Providers of DTC genetic tests claim that they provide many benefits, such as allowing consumers to prepare and watch out for diseases that they are at a high risk of contracting and enabling them to "make better lifestyle choices" (23andMe 2013a). However, there are many criticisms of and concerns about the tests in the literature.

One common concern is the effect that the disease-risk information generated by DTC genetic tests may have on consumers' health behaviour and health anxiety (c.f. Samuel et al 2010). Surprisingly, very few studies have tried to assess this. Indeed, to the author's knowledge, only three studies have examined the effects of the multiple disease risk information on a sufficiently large group of participants: Gordon et al (2012), Kaufman et al (2012) and Bloss et al (2011b). Although these three studies have provided useful information, they are also contradictory: Gordon et al (2012) and Kaufman et al (2012) found that some participants had changed their health behaviour after receiving their results, whereas Bloss et al (2011b) found no difference in participants' health behaviour pre- and post-test. All three studies also have potential biases. For example, Bloss et al (2011b) and Gordon et al (2012) sought out participants and provided them with a free or heavily-subsidised test; possibly preventing the generalization of their results to real consumers of an expensive new product. Kaufman et al (2012) avoided this by the use of participants who were actual consumers of DTC genetic tests; however, their participants were contacted through the providers of the tests and so independence cannot be guaranteed. The studies have further potential biases in their assessment of changes to health behaviour. For example, Bloss et al (2011b) only measured two health behaviours, thus excluding many other changes that consumers may have made. Although Gordon et al (2012) and Kaufman et al (2012) were open to all changes made, they relied solely on participants' correct recall; this can lead to issues such as recall or demand-characteristics bias. The contradictory nature and potential biases of these studies clearly show that the resolution of this issue is still a gap in the literature.

Another common concern relates to the information provision on the websites of providers of DTC genetic tests. Several studies have assessed their information content and criticised it as poor (c.f. Lachance et al 2010). There are concerns that this poor information content may affect consumers' understanding of the tests and their ability to give informed consent. However, to the author's

knowledge, no research has been conducted into information provision from the consumers' point of view. For example, no study has been found that has analysed consumers' information needs, consumers' information-seeking behaviour or the provision of information based on information that consumers themselves wish to know, rather than what professionals or the researchers think that they should know. There has also been no assessment of consumers' opinions about the information which the companies provide. This area is also, therefore, a considerable and important gap in the literature.

3 Background and Review of Information Behaviour

3.1 Introduction

This chapter describes information-behaviour research, examines a number of information-behaviour models and delves briefly into health information and health-information behaviour. It is included for two main reasons. Firstly, as described in the introduction, a major part of this research is related to consumers' information-seeking behaviour, whether it be their actual behaviour, their information needs or the information provided to them on the websites of providers. This part of the research is mainly exploratory (see Chapter 4) and so the inclusion of this chapter provides a background from which to think about consumers' information behaviour. Secondly, the fourteenth research objective (see section 1.3) involves the creation of a model which describes the main findings of the research in relation to consumers' information-seeking behaviour. The examination of a number of information-behaviour models in this chapter therefore provides a useful background to this model.

3.2 Background

Information behaviour is a term that "covers all aspects of people's dealings with information, including their opinions and judgements" (Robinson 2010, p.74), where information is defined as every instance when individuals "interact with their environment in any such way that leaves some impression on them" (Bates 2010, p.2381). Information behaviour includes "encountering, needing, finding, choosing, and using information"; behaviours which are fundamental to everyday life (Case 2006, p.4). It can also include concepts such as information avoidance, where an individual pursues behaviour that is intended to "prevent or delay the acquisition of available but potentially unwanted information" (Sweeny et al 2010, p.340).

Information behaviour has been studied for almost a century, during which time it has undergone an evolution in focus. Early research analysed the use of information sources with a focus on the sources themselves. Over time the emphasis changed to focus on the individuals who came into contact with the sources of information. Since the dawn of the internet the focus has continued to evolve, becoming "more integrated" and "less dictated by sources and institutions" (Case 2006, pp.4-6).

Information behaviour can be studied in three ways: inference, where an individual's information behaviour is estimated based on what the researchers know "about them and the context in which they need information", indirect study, where an individual's information behaviour is observed and

analysed, and direct study, where individuals are asked "what they need and what they do to get it" (Robinson 2010, pp.75-77). Research on information behaviour has looked at a wide array of topics from the information that "people glean from the mass media" to the information needs of scientists, doctors and engineers (Case 2006, pp.10-13).

3.3 Models of Information Behaviour

3.3.1 Background

There are a large number of different models of information behaviour. For example, a collection of models edited by Fisher et al (2005) contains 72 different models, theories and metatheories of information behaviour. Models are more tentative than theories, and "are most useful at the description and prediction stages of understand a phenomenon" (Bates 2005, pp.2-3), with the advantage of illustrating the approach the researcher has taken along with the most important "explanatory factors" that they have discovered (Case 2006, p.121). A number of models were examined, and those more commonly cited or considered appropriate are described in this section.

3.3.2 Wilson's Models

Some of the most interesting models of information behaviour are those developed by Wilson. Wilson developed several models over the years, which reflected the evolution and trends of the field. Two of his models are described here: his first model of information behaviour and his revised general model of information-seeking behaviour (Case 2006, p.123, 137; Wilson 2005, pp.31-36).

Wilson's first model of information behaviour was published in Wilson (1981). It was intended to describe the "fundamental categories of causal factors that produce" information need (Wilson 2005, p.31), and is shown in Figure 1.

The model shows 'information seeking behaviour' as the result of an individual's (or 'information user's') perceived information need (Wilson 1981). This need may originate from the individual, or may result from dissatisfaction with information that the individual already possesses (Case 2006, p124). After an information need has been perceived, the model shows three possible information-seeking actions for the individual to take. The first action is for the individual to "make demands upon formal [information] systems". These could include libraries and information centres. The second action is for the individual to make demands upon 'other information sources', which refers to systems that do not provide information as their primary function, but "may perform information functions in addition to" it. These could include estate agents or "car sales agencies". The final action the model recognises is for the information seeker to "seek information from other people" in

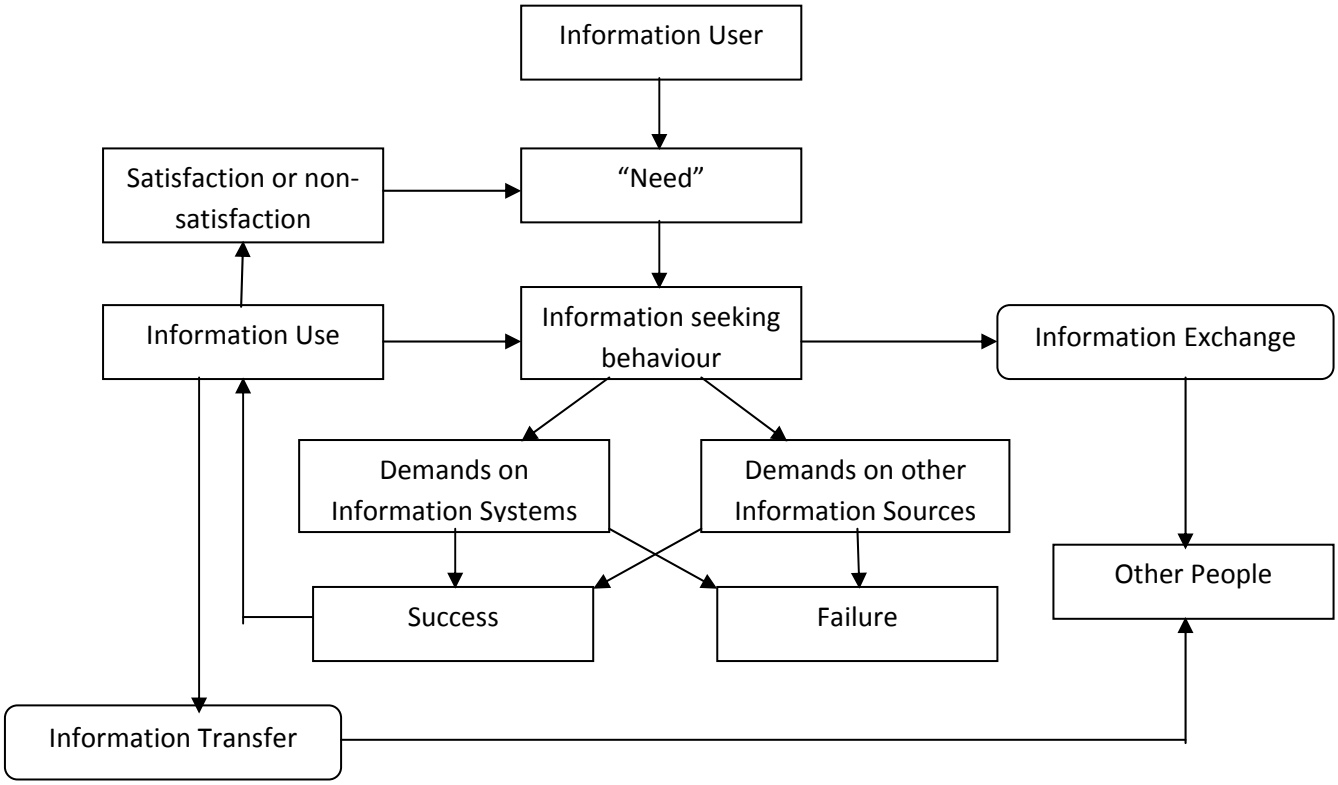


Figure 1 Wilson's first model of information behaviour (Wilson 1981, p.661).

an 'information exchange'; shown as a reciprocal behaviour in the model with the information seeker also transferring information to other people (Wilson 1981, pp.6, 7). This 'information exchange' is one of the key points that Wilson's model brought to attention (Case 2006, p.124).

The model shows the information-seeking activities resulting in either success or failure. If there is success, then the information is at some point used (given a wide definition to include activities such as evaluation of the information). That use may result in the information being considered satisfactory, may create new information needs or seeking if considered unsatisfactory, or may result in the transference of the knowledge to other people if thought useful to them (Wilson 1981).

Wilson's revised general model of information behaviour was first published in Wilson and Walsh (1996). It was developed from two of his previously published models, both from his original 1981 paper: 'the information use and the universe of knowledge' model and the 'information need and seeking' model (Wilson 2005, pp.31-33). Wilson's revised general model of information behaviour is shown in Figure 2.

This model is designed to "fill the gap" between an individual, or "person-in-context", and their decision to begin information seeking (Wilson & Walsh 1996). It "draws attention to the interrelated nature of theory" in the field of information behaviour and gives a general picture of information

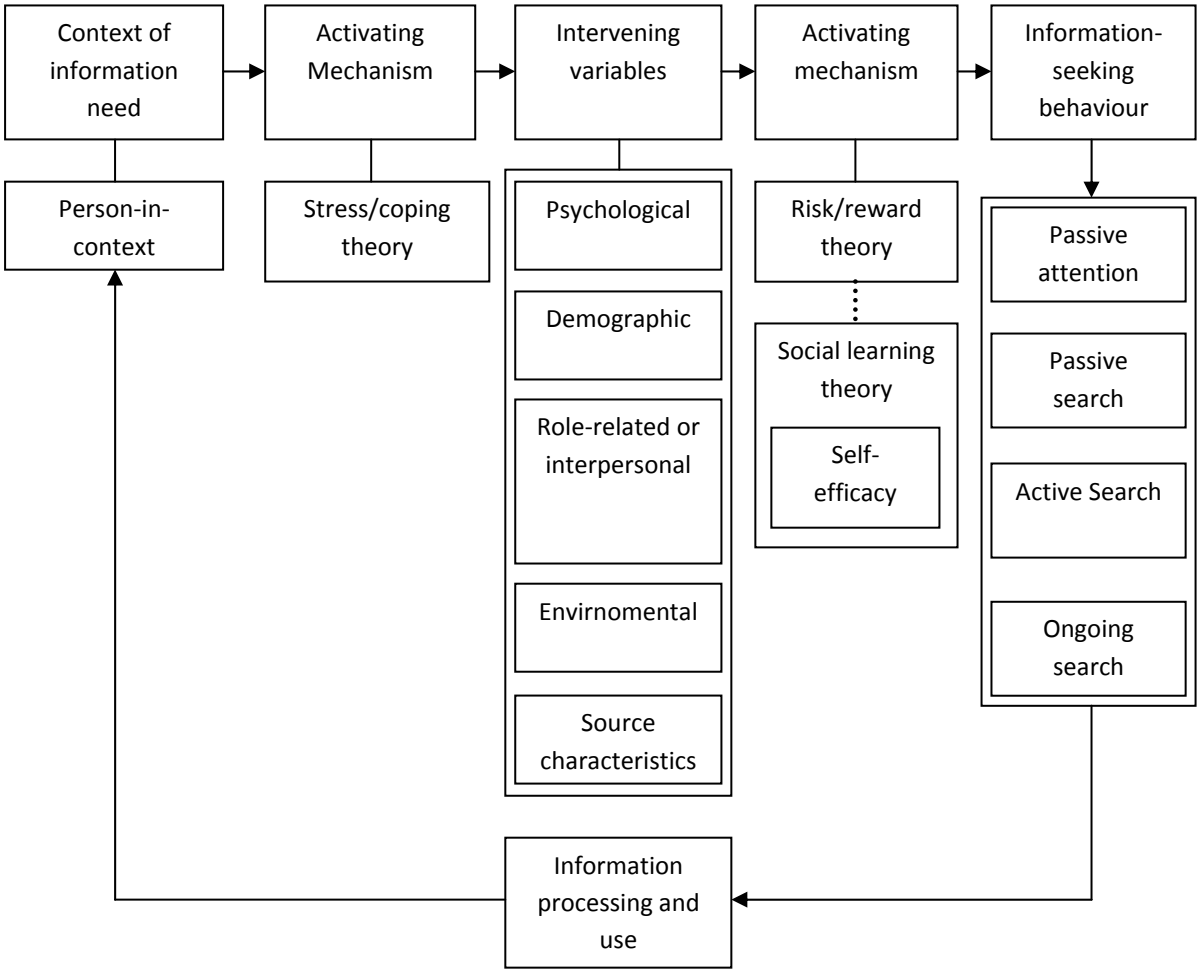


Figure 2, Wilson's revised general model of information (Wilson and Walsh 1996).

behaviour with aspects that are related to many different theories in the field. It is not a theory in itself but "a theoretical framework", with the aim of "linking theories to action". It thus allows a researcher to identify how their study fits into the whole (Wilson 2005, p.35). For example, 'stress/coping theory' is a psychological theory that explains why some information needs have a higher chance of causing an information-seeking process to occur than others. In the model this is connected to an 'acting mechanism', which could be considered as a "motivator". Therefore, these components represent the reason that someone searches for information. The other 'acting mechanism' is connected to 'risk/reward theory' and 'social learning theory'. 'Risk/reward theory' is a consumer research theory that explains "why some sources of information are used more than others", and 'social learning theory' is a psychological theory that explains the reason why a goal might or might not be pursued successfully based on an individual's self-efficacy (Case 2006, pp.136-137).

The 'person-in-context' component is connected to the 'context of information need'. This first component is a summary of Wilson's 'information need and seeking model', which includes factors such as environment, work role, physiological needs and personal barriers (Wilson 2005, pp.31-35).

The 'intervening variables' are barriers to the information-seeking process; many different types of possible barriers are included, such as 'demographic' and 'environmental'. The model also includes 'information processing and use', which represents "stages beyond information-seeking", providing a link back to the situation where an information need is first recognized (Wilson & Walsh 1996).

Finally, Wilson's model has four different types of information-seeking behaviour. This is an important part of the model as it includes types of seeking that are passive rather than active (Case 2006, p.137; Wilson & Walsh 1996).

3.3.3 Krikelas's Model of Information Seeking

One important early model is Krikelas's model of information seeking, shown in Figure 3.

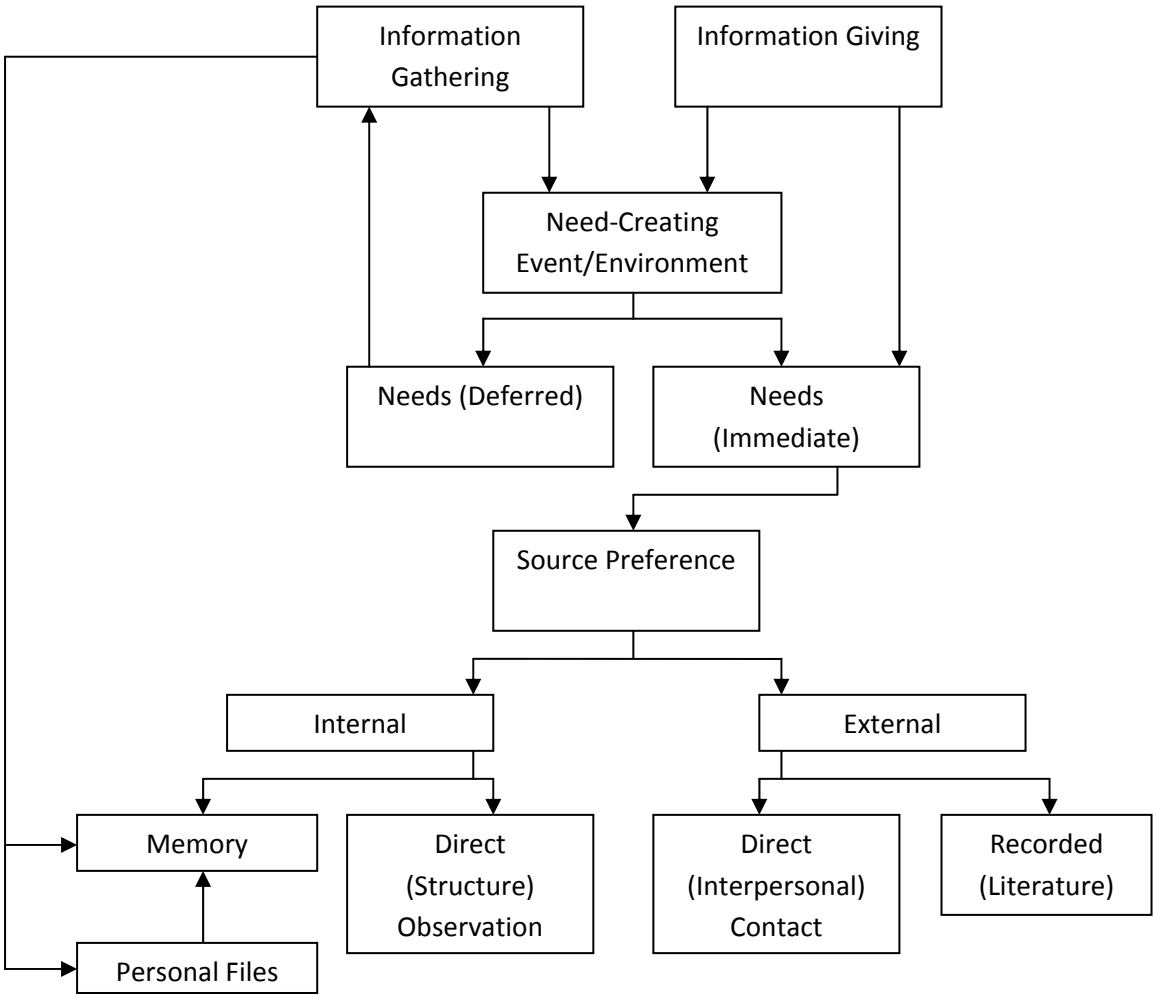


Figure 3 Krikelas's Model of Information Seeking (Krikelas 1983, p.17).

Krikelas's model is based on three different types of information activities: information giving, information gathering and information seeking (Henefer & Fulton 2005 pp.226-228). Information giving and information gathering are represented as individual components, and are located at the beginning of the sequence, and information seeking is split into several components further down the chain. The three information activities are mediated by information needs, which Krikelas (1983, p.10) defines as "a recognition of the existence of ... [an] uncertainty in the personal, or work-related, life of an individual". Unconscious needs are not included in the model, and needs are split into immediate and deferred needs (Henefer & Fulton 2005, p.226).

Krikelas (1983, p.13) defines information gathering as "those activities in which stimuli are accepted and held in storage to be recalled on demand". A good example of this type of activity is a researcher's reading of the literature to keep abreast of the latest developments in the field, rather than searching for a specific piece of information; another is an individual's general reading of the news. Such activities are not urgent, and hence Krikelas's model shows them as a result of deferred rather than immediate needs, and they are shown to be stored for future use in the individual's memory or personal files.

In contrast to information gathering, information seeking responds to an immediate need. The model shows that individuals will then choose their preferred source to find the information to fulfil it. This source could be either internal or external, with internal sources defined as those originating from the individual themselves i.e. memory, personal information storage (files) and observation, and external as those originating from other people i.e. interpersonal contact and recorded information (Henefer & Fulton 2005, p.226; Case 2006, p.126).

Krikelas (1983, p.17) defines information giving as "the act of disseminating messages", whatever form that information is in. It can be part of both the information gathering and information-seeking process, with the same act of information giving capable of being part of different processes for different individuals, or the same individual at different times (Krikelas 1983; Henefer & Fulton 2005, p.229).

Krikelas's model is often criticised as being oversimplified (Case 2006, p.124; Henefer & Fultun 2005, p.225). For example, although the model shows that information needs may be affected by information seeking (through their effect on the need-creating environment or by precipitating a need-creating event) there is no mention of the personal factors that may also influence them such as demographics, beliefs or ability. The same is true for 'source preference', where the model simply asserts that individuals will choose their preferred source without discussing factors that may

influence this. The model is also limited due to its bias towards information seeking in an occupational context (Case 2006, p.124; Henefer & Fultun 2005, p.225). However, it highlighted both the role of uncertainty as a motivator and the possibility that the information query may be answered by peers or by the memory of the seeker themselves (Case 2006, p.124). It can also be considered a "turning point" in user studies research due to the establishment of new research guiding criteria and as "groundwork" for future models and theories (Henefer & Fultun 2005, p.225).

3.3.4 Leckie et al's General Model of the Information Seeking of Professionals

Another interesting model is the general model of the information seeking of professionals derived by Leckie et al (1996), who based it on a meta-analysis of the relevant literature (Leckie 2005, p.159). Figure 4 shows this model.

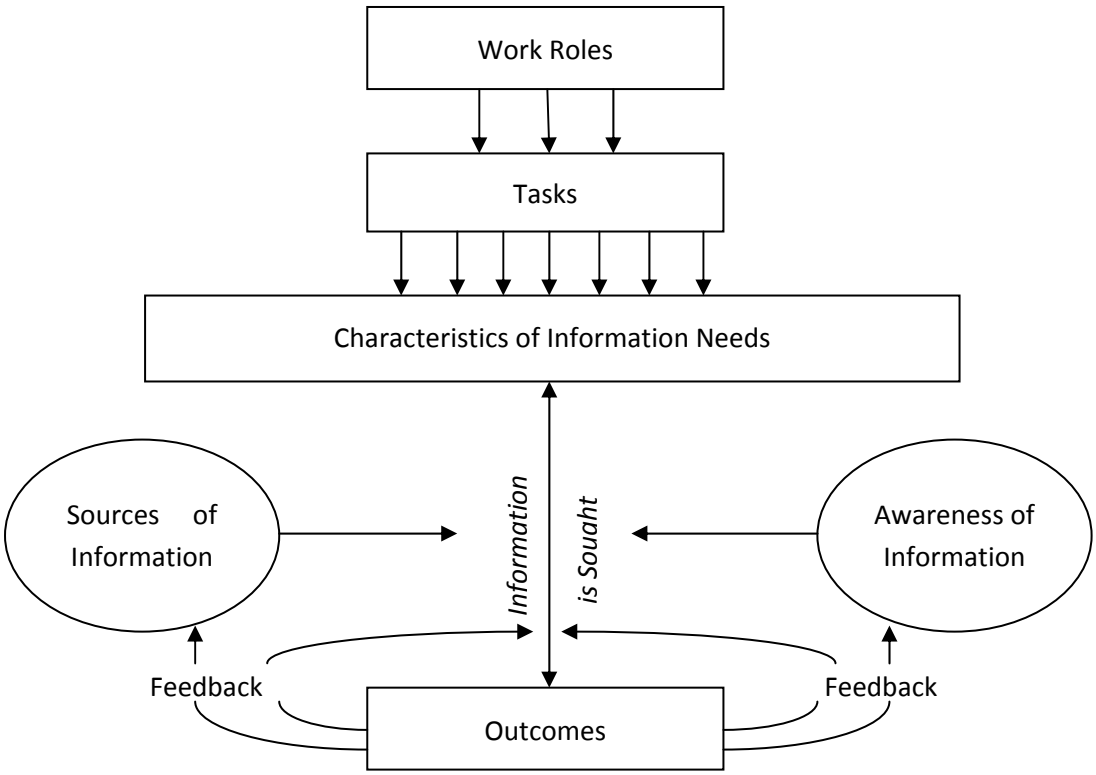


Figure 4 Leckie et al's (1996, p.180) General Model of the Information Seeking of Professionals

The model is based on their five main findings from the literature review. The first of these was that, even though professionals are normally trained for (and have expertise in) one particular area, a professional occupation normally consists of several "complex and different work roles". The second finding was that each role has "a constellation of tasks associated" with it and the third was that the tasks for each role are likely to create an information need and/or a requirement for

information seeking (Leckie 2005, p.159). It can be clearly seen that these three findings are represented by the top two components in the model: work roles and tasks. In the words of Leckie et al (1996, p.180) "the basic supposition of the model is that the roles and related tasks undertaken by professionals in the course of daily practice prompt particular information needs, which in turn give rise to an information-seeking process."

The fourth finding was that "intervening factors" may help or hinder both the information-seeking process and the use of the information (Leckie 2005, p.159). These factors include the information sources that are available and the individual's awareness of these sources, both represented by two of the remaining components in the model. The other set of intervening factors is represented by 'characteristics of information needs'. This refers to factors that affect the information need of the individual, such as demographic variables, occupation and location (Leckie 2005; Leckie et al 1996).

The final finding was that "more than one attempt" is often needed to fulfil an information need. This necessitated the inclusion of a feedback mechanism (Leckie 2005, p.160).

The slight vagueness of terms and the lack of any contextual factors in the model were deliberate in order to allow its use within a wide variety of work environments, with the intention that they would be filled in as appropriate (Leckie 2005, p.162). Although this recognition of the importance of contextual factors and in-built flexibility is an advantage of the model, it would perhaps benefit from some guidance as to how these factors could be incorporated. Another advantage is the inclusion of feedback loops, with the recognition that information awareness, knowledge of information sources, information seeking and the characteristics of the information needs can all be influenced by the outcomes of the information seeking process. However, tasks are shown as discrete entities, with no recognition that they themselves may be influenced by outcomes. Although limited by its applicability only to occupational situations (Case 2006, p.129), Leckie et al's model has been supported by various studies (cf. Landry 2006).

3.3.5 Bystrom and Jarvelin's model of information activity in work tasks of varying complexity

Another model that focuses on work-related information seeking is Bystrom and Jarvelin's model of information activity in work tasks of varying complexity. Although work-related, it does have the potential to be altered to analyse other situations. It was based on a combination of the existing literature and a thorough study into the information activities of a small group of civil servants (Case 2006, p.129; Byström 2005, pp.174-175). Figure 5 shows this model.

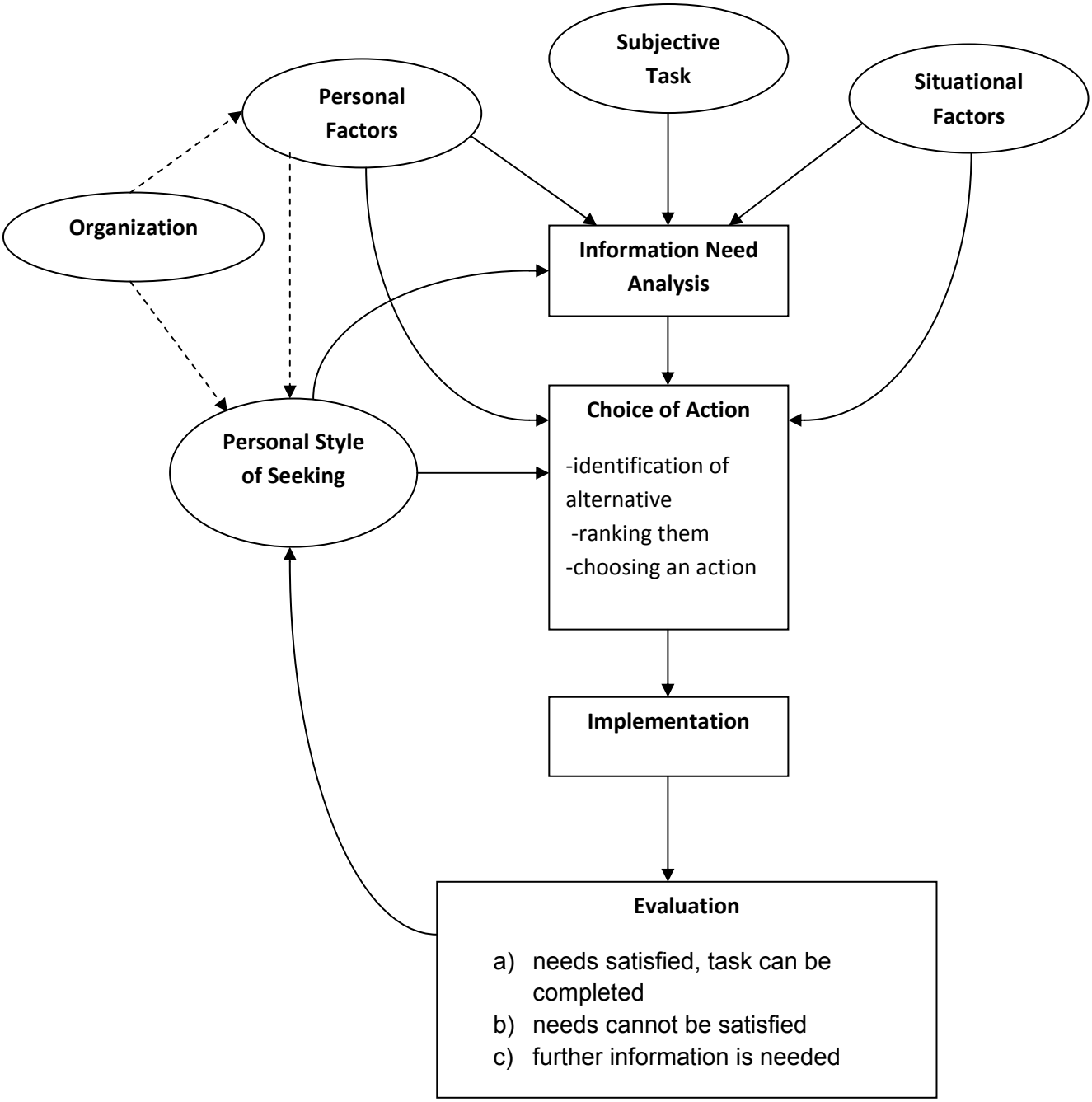


Figure 5 Byström & Järvelin's model of information activities in work tasks of varying complexity (Byström & Järvelin 1995, p.197)

One key aspect of Byström and Järvelin's research was the introduction of task complexity as a factor in the process of information seeking. Complex tasks are seen as those for which the individual does not know "exactly what needs to be done", and thus cannot be handled identically to routine tasks (Case 2006, p.129). Although task complexity is not represented in the model by a single component, Byström & Järvelin (1995, pp.196, 211) state that "a complex task may require several processes through the information seeking flow-chart", thus necessitating the inclusion of

the evaluation-based feedback loop, and that there was a relationship "between task complexity, the types of information needed [and] the number and types of sources and channels considered and used", decisions on which would be taken in the 'choice of action' component. As Byström (2005, p.176) states, the more complex the task the "more types of information" an individual must acquire, and the "less certain [an individual is] to predict what types of information are necessary to acquire".

In the model, a subjective task feeds into the information needs analysis. This analysis is affected by situational factors, such as the time available, and organization factors (Byström & Järvelin 1995, p.196), mediated in the model by personal factors. Personal factors and situational factors also affect the 'choice of action', but in this case refer to the individual's perception of the accessibility (either "cognitive, economic or physical") of the different information sources. The individual's personal style of information seeking is also a factor affecting both 'information need analysis' and 'choice of action' (Byström & Järvelin 1995), which is itself influenced in the model by organizational and personal factors.

As stated above, the model contains an evaluation-based feedback loop. The evaluation of the information found can be one of three options: the information has satisfied the information need, the information need has not been satisfied but can be by further information and the information need is not capable of satisfaction (Byström & Järvelin 1995).

The model benefits from the inclusion of the evaluation feedback loop, and the recognition that this process is mediated by an individual's personal style of information seeking. Another benefit is the inclusion of personal and situational factors as influences to both the information needs analysis and the choice of action. What is perhaps missing is a recognition of the effect that these factors may also have on the evaluation process itself.

3.3.6 Savolainen's Model of Everyday Life Information Seeking

Unlike Krikelas's, Leckie's and Byström and Järvelin's models, Savolainen's model of everyday life information seeking does not focus on work-related information seeking, but on information seeking which occurs as part of everyday life. This was not intended to create a sharp divide between work-related and non-work-related information seeking; Savolainen describes the model as complementary to work-related models. The model was tested by interviewing 11 teachers and 11 industrial workers (Savolainen 1995; Savolainen 2005, pp.143-147), and it is shown in Figure 6.

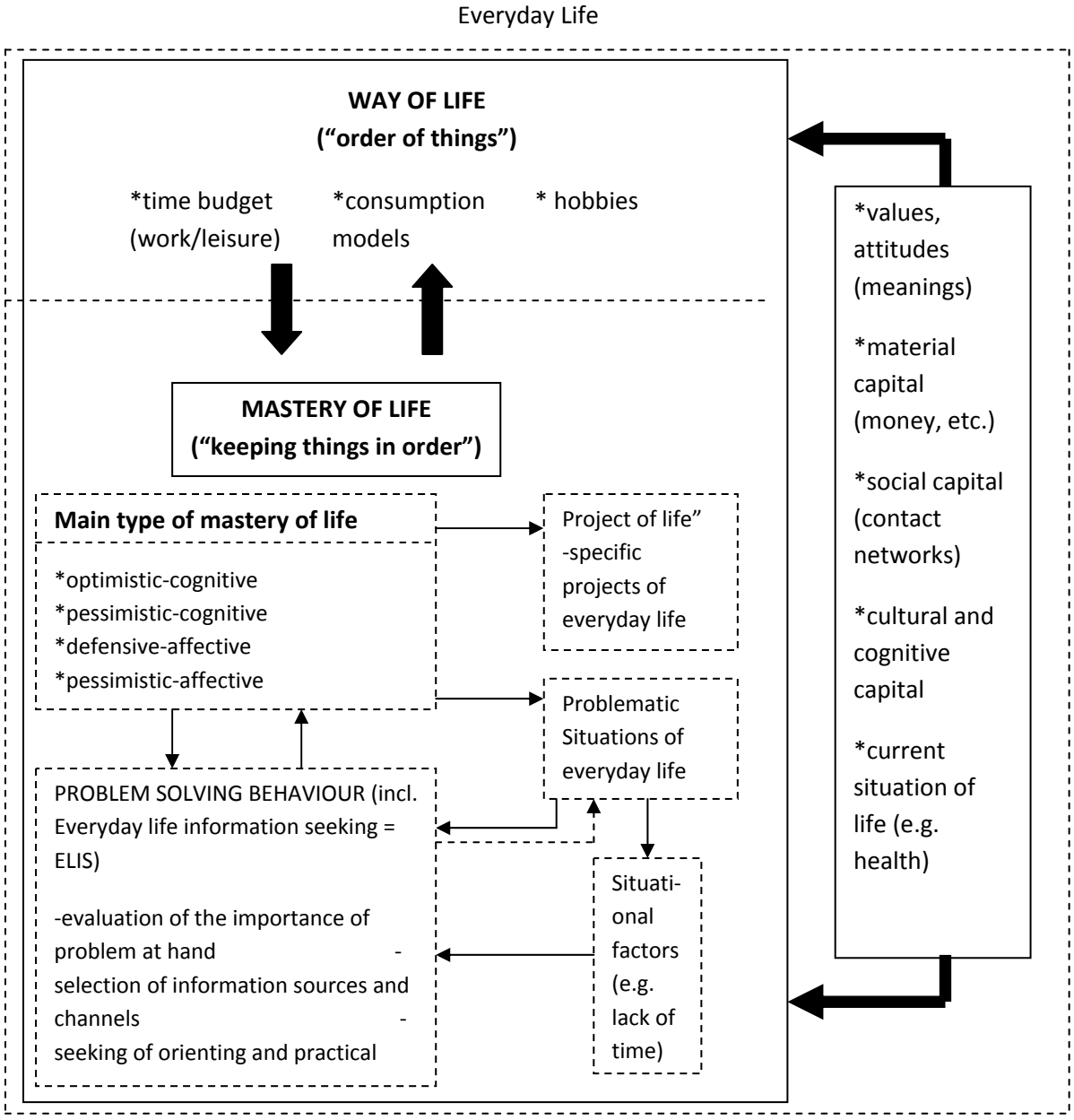


Figure 6 Savolainen's model of everyday life information seeking (Savolainen 1995, p.268)

A key point of Savolainen's model was the inclusion of sociological factors, such as social capital and wealth, in the information-seeking process (Case 2006 pp.131-132). As Savolainen (2005, p.143) states, the model's development was "primarily motivated by the need to elaborate the role of social and cultural factors that affect people's way of preferring and using information sources in everyday settings". It also encompasses a bigger time frame than many models, looking at more than information seeking in one particular time or situation (Case 2006, p.132).

Savolainen's model should be considered as less of a causal flow chart, and more of a presentation of factors and concepts affecting information seeking in ordinary life that should be explored in

depth in any relevant study (Case 2006, p.132). One of the main concepts is that of 'way of life'. 'Way of life' provides a context for the different factors which affect information seeking (Savolainen 2005, pp.143-144). It is defined by Savolainen (1995, p.259) as the "order of things", and represents those activities of life that an individual will do as part of their 'normal' life. It is the order that individuals have created "through their choices", one in which they have an interest to keep "as long as they find it meaningful" (Savolainen 2005, p.144).

A second main concept in the model is 'mastery of life'. This is shown as influencing, and being influenced by, 'way of life', and is described by Savolainen (1995, p.264) as the act of caring for and maintaining the 'way of life'. It involves solving problems (or disturbances in the 'way of life') in a pragmatic way and can be described as "a general preparedness to approach everyday problems in certain ways in accordance with one's values". When an information need appears then information seeking becomes an active part of the act of caring of the 'mastery of life' (Savolainen 2005, pp.144-146), eventually creating a set of "information-seeking habits" (Savolainen 1995, p.265).

The model gives several factors that influence both 'way of life' and 'mastery of life', including values, attitudes and 'social capital'. These represent the sociological factors that build up 'mastery of life', leading an individual to do things in their own, individual way, that is 'normal' for them. As stated above, solving problems pragmatically (including information problems) is part of the 'mastery of life', and the model gives the processes that an individual will do. These processes will be built up over time and influenced by all of the factors in the model (Savolainen 1995; Savolainen 2005, pp.143-147).

Finally, the model gives four different types of 'mastery of life', which influence (and are influenced by) problem-solving behaviours, and hence information seeking, both directly and indirectly. The first of these is 'optimistic-cognitive'. Individuals with this type have "a strong reliance on positive outcomes for problem solving". They believe that detailed analysis will solve most problems, and so will seek information from a wide variety of sources. The second is 'pessimistic cognitive', wherein individuals are "less ambitious" in their problem solving objectives. They may still be "systematic in problem solving", but will except that not all problems can "be solved optimally". The third type is 'defensive-affective'. In this type, as in 'optimistic-cognitive', individuals believe that detailed analysis will solve most problems. However, individuals of this type will avoid risk and so also may avoid situations where they must "actively seek information". The final type in Savolainen's model is 'pessimistic-affective'. In this type individuals will avoid problems, and do not use their abilities in an effort to solve any problems that do arise. Individuals of this type will avoid most instances of information seeking (Savolainen 1995, pp.265-266).

Savolainen's model is perhaps too rigid when describing types of mastery of life, with individuals divided into four distinct categories. However, overall the comprehensive description of how various sociological factors can affect information seeking in everyday life is an important contribution to the field.

3.3.7 Kuhlthau's Model of the Information Search Process

Kuhlthau's model of the information search process is based on a large amount of research over a timeframe of 20 years, including several longitudinal studies. It is focused on "intellectual access to information and ideas" and the processes by which people seek meaning from information (Kuhlthau 2005, p.230). Figure 7 shows this model.

The model shows the process of information seeking as one of "construction", and aims to describe the experience of information seekers "as a series of thoughts, feelings, and actions". Thoughts are shown as progressing from "uncertain, vague, and ambiguous" at the start of the information-seeking process, to more specific, clear and focused as the process moves forward. Feelings about the information-seeking process are those of "anxiety and doubt" at the beginning, moving to "confident and certain" towards the end. Actions begin as general information seeking, and end the process as more specific and focused (Kuhlthau 2013).

Kuhlthau's model splits the "thoughts, feelings, and actions" described above into six stages from the beginning of an information-seeking task to the end (Kuhlthau 2005, p.230). These are shown at the top of the model and are: 'Initiation', 'selection', 'exploration', 'formulation', 'collection' and 'presentation', leaving a final stage of 'assessment' at the end of the process.

'Initiation' is the start of the information-seeking process, when an individual becomes aware of an information need. This can often cause a sense of anxiety or uncertainty. In this stage the problem is contemplated and possibilities for solving it discussed. The second stage, 'selection', is the identification of the general information-seeking area. In this stage the uncertainty that arose in the 'initiation' stage will often change to optimism. The individual will think about which topics to pursue, influenced by factors such as task criteria and information and time available. Advice may be asked and information topics may be scanned (Kuhlthau 2005, pp.230-231; 2013).

The third stage, 'exploration', is where information is sought about the general area identified previously (Kuhlthau 2013). Normally "inconsistent, incompatible information is encountered", which can cause levels of "uncertainty, confusion, and doubt" to increase (Kuhlthau 2005, p.231). The information seeker will aim to discover enough information to form their own viewpoint, and

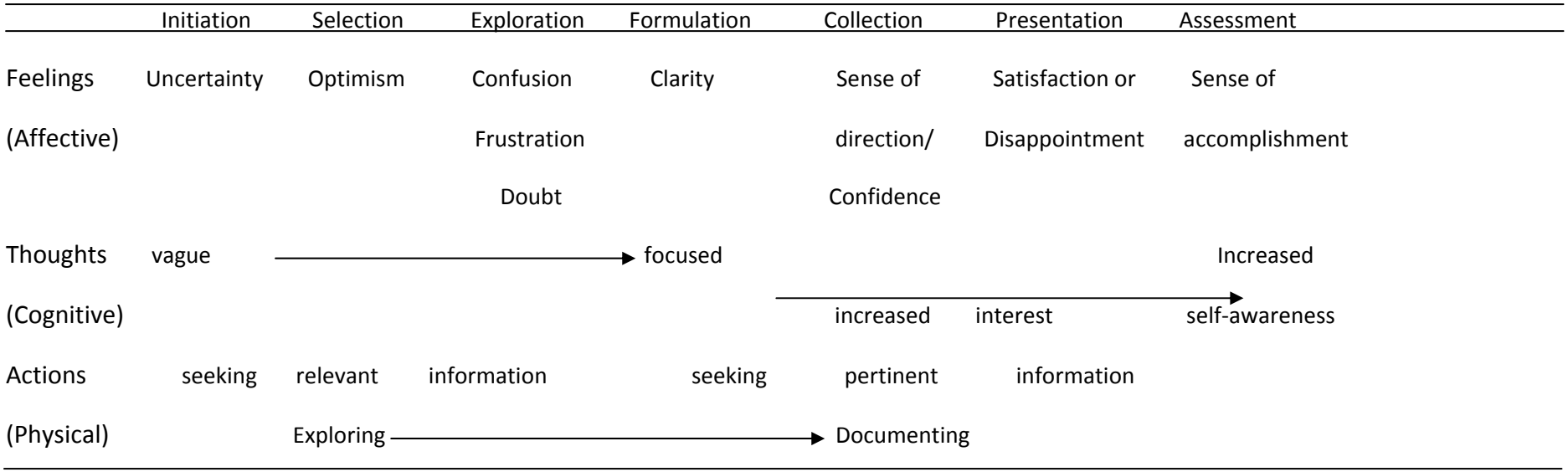


Figure 7 Kuhlthau's model of information seeking (Kulthau 2013)

will read “to become informed” about the general area, fitting information they find to what they already know. This stage is the most difficult of the information process, and one in which the search may be given up. Individuals may be discouraged or threatened by the situation, and it may cause “a sense of personal inadequacy” (Kuhlthau 2013).

‘Formulation’ is the fourth stage of the model, and is where “a focused perspective is formed”. This normally causes a reduction in uncertainty and an increase in confidence (Kuhlthau 2005, p.231). Ideas are selected and identified, and the area “becomes more personalized”, with individual’s gaining an insight into the topic and a sense of meaning (Kuhlthau 2013).

The fifth stage, ‘collection’, is where the “interaction between the user and the information system functions most effectively and efficiently”. Relevant information is selected and gathered, and detailed notes of it made. General information is ignored and pertinent information sought. The information seeker will continue to increase in confidence and decrease in uncertainty (Kuhlthau 2013).

The final stage of the model’s information-seeking process is ‘presentation’. In this stage the information seeking is completed and the information is prepared to be put to use. Individual’s will often feel relief at this stage, and either satisfaction or disappointment depending on how well the search has gone (Kuhlthau 2005, p.231 and 2013).

One of the key contributions of Kuhlthau’s model is the inclusion of “affective aspects or feelings in the process of information seeking” (Kuhlthau 2013), and the identification of the point in the ‘initiation’ stage where confidence drops and uncertainty grows (Kuhlthau 2005, pp.231-232). However, no attention is paid to how an individual’s personal factors may influence the various components in the model.

3.3.8 Dervin’s Sense-Making Methodology

Dervin’s sense-making methodology focuses on the relationship between information, communication and meaning, and is important in “understanding how human beings derive meaning from information” Tidline (2005, p.113).

Sense-making was originally based on constructivism, wherein it was “assumed that all information is simply the sense made by individuals at specific moments in time-space” (Dervin 1983). More recently it has included aspects of communitarism, with the idea that sense-making behaviours allow an “individual to construct and design his or her movement through the time-space context” (Savolainen 2006, p.1117). The focus of the methodology is on the process by which people use

observations, both their own and those of other people, in order to create "pictures of reality" which guide their behaviour. Since it is assumed that humans have a large number of gaps in their view of reality, and that information seeking and communication is "gap-bridging behaviour", sense-making behaviours are thought to be frequent. Indeed, Dervin states that "each new moment in time-space requires another gap-bridging step" (Dervin 2005, pp. 26-28). Sense-making behaviours are assumed to be "potentially responsive" to changing conditions or situations (Dervin 2005, p.27), and so sense-making models use "changing situations as predictors", rather than demographics or characteristics of the individual. Sense-making research is focused on the "situational conditions" that cause particular sense-making behaviours, with an emphasis on patterns rather than "mechanistic input-output relationships" (Dervin 1983).

The original model proposed by Dervin for sense-making research was the situations-gaps-uses model, where situations are the "time-space contexts at which sense is constructed", gaps are the information needs of individuals, or the questions they have as they "construct sense and move through time-space" and uses are the effect the information has on the individual or the uses to which they put "newly created sense" (Dervin 1983). A common data collection method using this model is the time-line interview, where participants are asked to give a step by step description of their sequence of information seeking (Tidline 2005, p.113; Dervin 1983).

Over time sense-making moved to a more holistic approach with a stress on "verbing" (Tidline 2005, p.114). Verbings are elements that are used by individuals as part of the sense-making process, such as hunching, defining and factizing, and are important to the sense-making principle that there is not just "one right way to produce knowledge or to use information" (Savolainen 2006, p.1117). The model also expanded to include context, such as cultures and "power structures", outcomes, such as helps, functions and consequences, and bridge, such as ideas, attitudes and feelings, alongside situation, verbings and gap (Dervin 2005, p.28).

3.3.9 Summary

This section has presented various different models of information behaviour. As stated in section 3.1, a large part of this research is an exploratory investigation into consumers' information-seeking behaviour, including the creation of a model of the findings (see section 1.3). This section, therefore, provides a useful background from which to think about the relevant issues and there are several important points that should be taken from it.

Perhaps the most important point is the concept of information need. Information need can be defined as the recognition that one's "knowledge is inadequate to satisfy a goal" that one has (Case

2006, p.5) and several of the models include it as the cause of an individual's information seeking. Information needs are shown as influenced by various factors, such as the current task (Leckie et al 1996) the "satisfaction or non-satisfaction" of information found (Wilson 1981, p.661) and personal or situational factors (Byström & Järvelin 1995).

A second important point highlighted by several models is that information seeking does not occur in isolation. Rather, it can be influenced by factors such as psychology, demographics and environment (Wilson and Walsh 1996), an individual's preferred source (Krikelas 1983), their "awareness of information" (Leckie et al 1996, p.180) and their "personal style of seeking" (Byström & Järvelin 1995, p.197).

A third point is that information seeking is a process rather than a single event. Information seeking can involve the choice of an appropriate action, the implementation of the action and an evaluation of the information found (Byström & Järvelin 1995). It can be passive, active or ongoing (Wilson and Walsh 1996). The information found can form a feedback loop with the information needs, the individual's awareness of the information area and the sources chosen for the search (Leckie et al 1996). The act of seeking information can follow a predictable emotional pattern, from uncertainty to optimism, confusion, clarity, confidence, satisfaction and finally accomplishment (Kulthau 2013).

3.4 Health Information

3.4.1 Introduction

As stated in section 3.1, a large part of this research is related to consumers' information-seeking behaviour. Specifically, it is focused on information-seeking behaviour that is related to DTC genetic tests. Since DTC genetic tests are products which provide health information, it follows that much of the information sought relates to either health information or information about a healthcare product.

This section was therefore included to provide a brief description of health information and studies of health-information behaviour. The intent was not to provide a framework for the research, since it was considered that this might cause undue influence to research of a highly exploratory nature (see Chapter 4), but to provide a background to it.

3.4.2 Background

Information about health and healthcare is amongst the oldest type of information in the world, with recorded information dating back as far as the Bronze Age (Robinson 2010 pp.39-40). The

quantity of published health information has undergone a large growth since the start of the scientific era; Arndt (1992) estimates that there has been an exponential growth rate in the number of published biomedical articles and journals since 1750. Although this estimate may not be entirely accurate, it does show the exceedingly large quantity of published health information available (Robinson 2010 p.20). For example, Medline (the U.S. National Library of Medicine's main bibliographic database) contains more than 19 million references, with 700,000 added in 2010 alone (NLM 2013). Robinson (2010 pp.20-21) states that the quantity of information is such that no individual can "have a full knowledge of even a small area" and so external aids such as books and databases are essential.

Another area of rapid growth has been consumers' use of the internet to find healthcare information (Cline & Haynes 2001). For example, it was estimated that in 1997 almost half of American users of the internet had searched for health information (Eng et al 1998). In 2001, the Pew Internet and American Life Project estimated that 61% of American users of the internet had searched for health information (Pew 2001) and in 2012 that 72% had done so in the past year alone (Pew 2012). Consumers have no shortage of health information on the internet; a Swiss (and UN-accredited) NGO named the Health on the Net Foundation estimates that there are four billion pages on the internet that contain health information (Robinson 2010, p.22; HON 2013). Although this may be a slightly tongue-in-cheek estimate, it does illustrate the large volume of health information available on the internet.

3.4.3 Studies of Health-Information Behaviour

3.4.3.1 Background

According to Robinson (2010, p.77), health information has been the focus of more information-behaviour studies than any other area, with numerous studies focused on healthcare professionals, providers, carers, patients and the public.

One early paper on health information was by Brodman (1974, pp.67,70), who described and attempted to explain the change in the availability and usage of health information, and in particular medical libraries, from the sole preserve of a physician "demi-god" to a resource for ordinary patients. She described how changes in funding, access to and experience of healthcare, changes in patient attitudes towards doctors, greater public education in science, changing attitudes of politicians towards healthcare and medical research and the diversification of jobs in healthcare all contributed to this change. She also briefly delved into the information behaviour of users of medical libraries, stating that the majority of individuals will normally only seek an answer to specific

questions when visiting a medical library, and will use it as a last resort. Before visiting they will seek to elicit the information from peers, in particular the person in their group who normally has a good record in helping in such situations, the so-called "gatekeeper". In contrast, Brodman stated that researchers will also visit a medical library to find specific answers, but they will also visit to keep abreast of the literature in their field.

Another early study of health-information behaviour was conducted by Hibberd and Meadows (1980, p.169), who surveyed hospital doctors to investigate how they used sources of drug information. They found that one commercial source (the Monthly Index of Medical Specialities) was used most commonly for the majority of queries about the prescription of drugs and for learning about the existence of new drugs. However, sources published by the medical profession were more commonly used for finding out about the efficacy of new drugs. They also pointed out that no source available at the time met "all the needs of the prescribing doctor" and gave recommendations to remedy this.

After 1990 the volume of health-information-behaviour research greatly increased. One common area of study has been the information behaviour of health professionals (Robinson 2010, p.77). For example, Kostagiolas et al (2012) investigated the health information behaviour of psychologists in Greek Hospitals, finding that their most common information need was to do with psychotherapeutic interventions and their most commonly-used information source was personal libraries. Andrews et al (2005) investigated the information-seeking behaviours of health professionals in the Kentucky Ambulatory Network. They found that 50% used the internet at least a few times a week to search for drug information; 61% used a print reference source at the same frequency. Hider et al (2009) investigated the information-seeking behaviour of clinical staff in New Zealand. They found that nursing and allied health staff were less likely to use a search engine at least once a week than dental or medical staff, and that Google was the most-used electronic resource by all groups.

Other studies in this area have included research into the public as health consumers and the use of information-behaviour models in health-information research. These areas are described in the next two sections.

3.4.3.2 The Public as Health Consumers

As described above, one common area of health-information-behaviour study has been research into the public as health consumers. In this context, the term 'public' is used to represent anyone

who is not a health professional, whether they be a patient, carer or ordinary member of the general public.

One topic of this research has been examining patient's health-information behaviour. According to Case (2006, p.295), in recent years there has been a growth of interest in this, mainly caused by a combination of a general increase in patients' interest in health, an increase in patients' interest in preventative medicine, the growth in homeopathic and self-help books and the vast increase in health-information websites aimed at the lay person. One study in this area was by Attfield et al (2006), who compared NHS patients' pre- and post-consultation information needs. Their results showed that pre-consultation participants sought information about whether or not they (or a peer) actually needed to have a consultation (in order to avoid wasting either their time or that of the NHS) and where to go for the best possible consultation. After the arrangement of a consultation, participants tended to research background health information (i.e. about conditions and treatments) in order to both contribute during the consultative process and to have a knowledge base to properly judge the consultant's proposals. After a consultation, participants' information needs changed to one of research and confirmation of diagnosis, alongside the efficacy and safety of treatments.

Another topic of this research has been investigations into why individuals do not change their behaviour when presented with information encouraging them to do so (Case 2006, pp.295-296). This issue was investigated as far back as 1947, when Hyman and Sheatsley (1947, p.412) suggested that reasons included that "interested people acquire more information than the uninterested", that "people seek the sort of facts which are congenial to their existing attitudes" and that "different groups interpret the same information differently". More recently, Sligo and Jameson (2000, p.865) investigated why Pacific island immigrants to New Zealand had a lower than average uptake of cervical cancer screening, even though they had a higher than average risk of it. They found that barriers included "imperatives of cultural topic avoidance, modesty, and religion". It was also found that respondents preferred information to be mediated through their community, but that individuals performing the examination would not be of the same ethnicity. Enwald et al (2012) examined the information behaviour of pre-diabetic individuals in Finland. They found that participants with lower levels of physical fitness and higher values for their body mass index were keener than others to have access to individually-tailored information about physical activity and nutrition.

A third topic has been research into the use of the internet for health information. Cantrill et al (2005, p.1467) investigated the use of the internet for health information amongst adolescents in

the UK and the USA. They found that, in general, participants regarded health information on the internet as salient, which "was increased through active searching and personalisation". They concluded that the internet was a useful source for adolescents in regard to health information. Sillence et al (2005, p.401) reported on a large survey of internet users that investigated their health-information behaviour. Motivations for searching for health information online included "the quest for information or a desire to be in better control of one's health". The most-commonly-searched for topic was alternative medicine, followed by diet, women's health and cancer. This contrasted with an earlier survey in 2000 where cancer was the main topic searched for, followed by alternative medicine, diet and women's health. A wide range of different websites were used, with 250 mentioned in the survey. Concerns about the public accessing sub-standard health information on the internet are commonly articulated by health professionals. A study by Gunter et al (2004, p.375) found that, although there are concerns amongst professionals about the quality of some health information on the internet, and although 45 percent of participants stated that they had seen misleading health information when searching the web, the general public has "a healthy dose of scepticism" that should provide some reassurance.

3.4.3.3 Information-Behaviour Models in Health-Information Research

3.4.3.3.1 Background

As stated above, there have been a large number of studies of information behaviour in relation to health information. However, very few of these studies have been based on health-behaviour models (Robinson 2010, pp.74-75).

One example of this type of research is a study conducted by Beverley et al (2007, p.27). Beverley et al investigated the information behaviour of visually-impaired people in relation to health information and social-care information, and compared the applicability of Wilson's revised model of information behaviour and Moore's model of social information need. They found that both models were useful in the analysis and interpretation of the study data. In particular, Moore's "six dimensions of social information need" were well supported by the study, and his "clusters of information needs" were recognized and could be assembled into a hierarchy of levels of importance for people with visual impairments. Although Wilson's model needed to be slightly modified to apply to a specific rather than general group, his "intervening variables" accounted for some aspects not accounted for by Moore's model.

Another relevant study was conducted by Williams et al (2003). Williams et al used Dervin's sense-making model to investigate the low usage of an NHS information kiosk by women aged between 55

and 74, utilising a 'time-line interview' technique' to do so. They found that the study methods allowed them to identify many reasons for the low usage of the kiosk, including doctors' remaining function as the primary information provider, a lack of awareness of the availability and usefulness of the kiosk and a preference for knowing only the minimum necessary information for their condition.

3.4.3.3.2 Johnson's Comprehensive Model of Information Seeking

One important health-related model of information-seeking behaviour is Johnson's comprehensive model of information seeking (CMIS). Partly based on the health belief model (Johnson & Meischke 1993, p.343), CMIS was developed by Johnson with a focus on cancer-related information (Case 2006, p.133). Figure 8 shows this model.

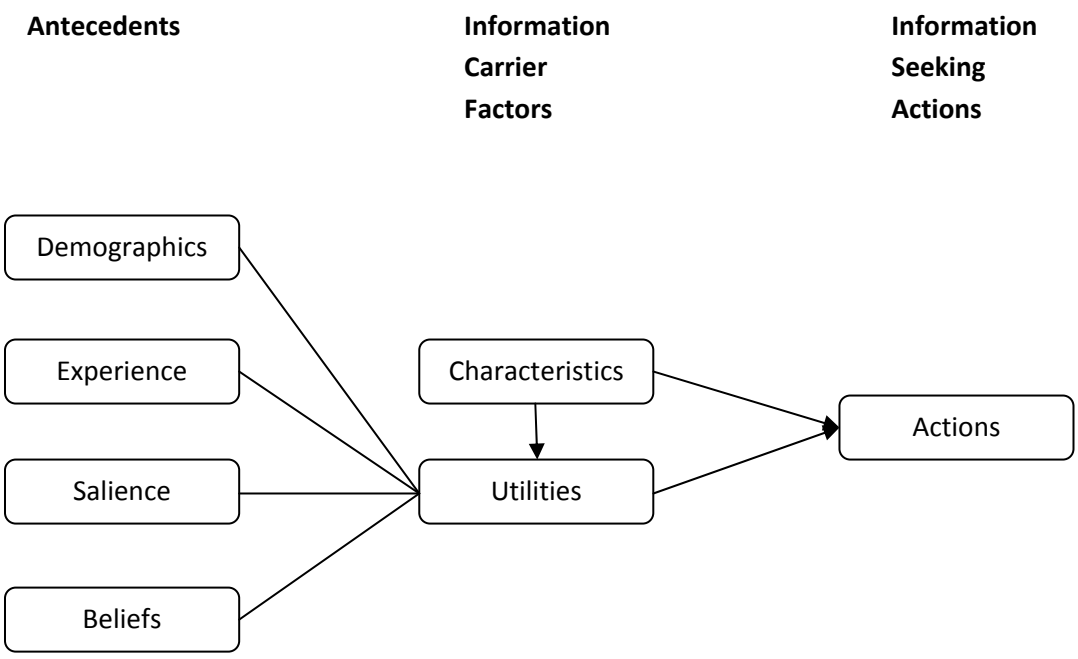


Figure 8 Johnson's comprehensive model of information seeking (Johnson et al 2001, p.340)

The model contends that "information-seeking actions" are determined by four health-related antecedents and two information carrier factors (Johnson & Meischke 1993, p.343). The four antecedents are factors which influence an individual's motivation to and likelihood of seeking information and their "natural predispositions" for how to do so (Johnson et al 1995, p.278). The first two antecedents are described as background factors and include demographics, which highlights the influence that factors such as age, socioeconomic status and ethnicity may have on an

individual's information seeking, and experience. Experience, also known as direct experience, describes how an individual's personal experience with an issue may affect their information seeking; for health-related information seeking this may refer to experience with a disease (such as cancer) and can include both an individual's direct experience (e.g. if they are themselves suffering from the disease) or the experience of an individual whom they know. This antecedent can include many different facets which can influence information seeking such as the stage of the disease, decisions about treatment and side effects (Case 2006, p.133; Han et al 2010; Johnson et al 2001). The third and fourth antecedents, salience and beliefs, are both factors described as personal relevance. Salience refers to the need for information to be applicable to the individual's situation and for them to consider it to be personally significant. The salience of information can affect an individual's motivation to search for information, and can be affected by factors such as their perception of their own disease risk. Beliefs refers to an individual's beliefs about any topic relevant to the issue. For example, it can include beliefs about disease and treatment, beliefs about what can be done to solve problems and beliefs about the individual's ability (Case 2006, pp.133-134; Johnson et al 2001; Han et al 2010).

The information and carrier factors are the characteristics and utility of the sources of information, or 'communication channels', which individuals use when searching for information. The characteristics of an information source include factors such as the type of source, whether it is interpersonal or mediated and an individual's preferences. Utility refers to how useful an individual believes or expects a source to be and how likely it is to satisfy their information needs. An individual's view of the utility of a source is shown in the model to be influenced both by their perception of the characteristics of a source and by the four antecedents described previously (Case 2006, pp.134-135; Johnson et al 2001; Han et al 2010).

The final section, information seeking actions, refers to the choices made and type of information-seeking behaviour undertaken, such as the extent of the search, methods used and sources chosen, which are, as stated previously, determined by the antecedents and information carrier factors (Case 2006, p.135; Johnson et al 2001).

The model is an important contribution that highlights many of the factors that may influence an individual's information-seeking behaviour, especially when searching for health-related information. However, it fails to include any feedback mechanisms or recognise how the information-seeking actions themselves may influence the other components in the model. It could also be considered to place too much reliance on the utility of information sources with some

research, for example, finding that accessibility can be a more important factor in some cases (Case 2006, p.135).

3.5 Summary

Information-behaviour research examines individuals' interactions with information, including their information needs, information seeking and use of information. There are a large number of models of information behaviour, from general to highly specific. Key points useful in information-behaviour research include the concept of information need (c.f. Leckie et al 1996, Wilson 1981 and Byström & Järvelin 1995), that information seeking does not occur in isolation (c.f. Wilson and Walsh 1996 and Krikelas 1983) and that information seeking is a process rather than an event (c.f. Kulthau 2013 and Wilson and Walsh 1996).

Health information is one of the oldest types of recorded information and one that is expanding exponentially. Studies of health-information behaviour can focus on patients, providers, healthcare professionals, carers and the public. Although it is one of the most studied areas of information behaviour, only a small proportion of health-information-behaviour research has been based on health-behaviour models.

4 Research Methodology

This chapter describes the research methodology. It begins by describing the methodological framework of the research, including the research paradigm and research design. It then briefly describes the methods of the previous studies in the research area, and finishes by giving the individual methods for the three studies (the survey, email interviews and the content analysis) which comprise this research.

4.1 Methodological Framework

4.1.1 Research Paradigm

4.1.1.1 Background

The term paradigm is a perhaps overused word that was originally conceptualized by Kuhn (Gauch 2002, p.84). One definition of a paradigm in science is "the broad common ground and disciplinary matrix that unites particular groups of scientists at particular times" (Gauch 2002, p.84). In terms of research design and analysis, the theoretical paradigm is the interpretative framework through which the research is viewed. Different theoretical paradigms are composed of different assumptions about aspects of research. These can be grouped into four areas: ontological, epistemological, axiological and methodological. The assumptions and hence the dominant paradigm commonly vary between research disciplines and communities. Individual researchers may change their assumptions over the length of their careers, and may use different paradigms for different studies (Creswell 2013, pp.6, 16-24). There are many different theoretical paradigms; those commonly used include positivism, post-positivism, constructivism/interpretivism and critical theory (Creswell 2013, pp. 15-39; Crotty 1998, p.5; Guba & Lincoln 2005, 191-195).

The paradigm that most closely fits this research is post-positivism. Post-positivism can be described as the traditional scientific method (Creswell 2009, p.6), with a logical, reductionist and "cause-and-effect" approach (Creswell 2013, pp.23-24), and an objective method of inquiry (Creswell 2013, pp.23-24; Creswell 2009, p.7; Guba & Lincoln 2005, p.195). Post-positivists believe that knowledge should be based on "data, evidence and rational considerations" (Creswell 2009, p.7) and that different participants will have different perspectives on reality (Creswell 2013, pp.23-24). They believe that outcomes are generally determined by specific causes, and reduce complex ideas into smaller testable ones (Creswell 2009, pp.6-7). Post-positivists emphasize reliability, validity and rigour, and see their position in policy debates as informers, rather than the critical-theory advocates or the constructivist facilitators (Guba & Lincoln 2005, p.196).

4.1.1.2 Positivism and post-positivism

In order to fully understand post-positivism, it is necessary to examine the related paradigm of positivism, from which it evolved. Positivism, a term popularised in the 19th century by Comte, refers to knowledge that is gained not by speculation but by "direct experience": experimentation and observation (Crotty 1998, pp.20-22). Comte believed that knowledge has historically passed through three different conditions: "the Theological, or fictitious; the Metaphysical, or abstract; and the Scientific, or positive" (Comte 1973, p.25), and it is the latter that he sought to promote (Crotty 1998, pp. 20-23). Comte saw himself as passing on a tradition that began with Bacon, and it is not controversial to argue that this emphasis on observation and experimentation, as well as the generation of scientific hypotheses and laws that followed, was a great improvement on science's previous reliance on the authority of ancient thinkers (Crotty 1998, p.23; Vickers 1987, pp. 1-5; Henry 2008, p.1).

As stated above, positivism is based on experimentation and observation rather than speculation (Crotty 1998, p.20-22). It is objective, and relies mainly on quantitative methods (Guba & Lincoln 2005, p.195). Research is conducted with the aim of testing theories and providing data for the development of scientific laws (Bryman 2008, p.14), which are aimed to be universal and causal (Robson 2002, p.20).

Post-positivism can be considered an "attenuated form of positivism" (Crotty 1998, p.29). The central difference between the two paradigms is their viewpoints on the nature of reality and scientific knowledge. Positivism argues that there is a single real reality that is able to be apprehended, but that only the scientific method can truly do so. In positivism, scientific knowledge is objective, accurate and true, and positivists aim to verify hypotheses. In contrast to this, post-positivism argues that although there is indeed a single real reality, it cannot be perfectly apprehended by any means. In post-positivism, scientific knowledge can only be probable and approximate, and cannot be entirely objective. Recognising this inability to determine absolute truth, post-positivists will always aim to disprove, rather than verify, a hypothesis (Guba & Lincoln 2005, pp.193-196; Crotty 1998, pp.26-34; Creswell 2009, p.7). This evolution in viewpoints occurred during the 20th century (Crotty 1998, pp.29-34). However, the beginnings of the idea can be traced back at least as far as Descartes, who argued that (excluding divine explanations) the only truth that can be absolutely known is that one exists (Descartes 2008).

4.1.1.3 Comparison with other paradigms

As described above, the assumptions of which the different paradigms are composed can be grouped into four areas: ontological, epistemological, axiological and methodological (Creswell 2013 pp. 19-21).

Ontological assumptions deal with "the nature of reality and its characteristics" (Creswell 2013, p.20). As described above, positivists consider there to be a single real reality that is able to be apprehended; post-positivists agree that there is a single real reality, but think that it cannot be perfectly apprehended (Guba & Lincoln 2005, p.195). In contrast to this, constructivism/interpretivism is based on the idea of relativism (Guba & Lincoln 2005, p.195). In this viewpoint there is not an objective reality, but rather multiple subjective realities created through individuals' interactions and experiences (Robson 2002 pp. 22-28; Creswell 2013 p.36; Guba & Lincoln 2005 p.195). Crotty (1998, p.42) states that in this paradigm "meaning is not discovered but constructed". Unlike constructivism/interpretativism and similar to positivism and post-positivism, critical theory is realist rather than relativist (Guba & Lincoln 2005, p.195). However, critical theorists see this reality as having been shaped by historical ideology, such as views on gender and race, and the difference in power between various groups in a society (Guba & Lincoln 2005, p.195; Creswell 2013, p.36; Crotty 1998, p.157). As Crotty (1998, p.157) states, in critical theory "facts can never be isolated from the domain of values or removed from ideological inscription".

Epistemological assumptions deal with "what should pass as acceptable knowledge" (Bryman 2008, p.693) and "how reality is known" (Creswell 2013, pp. 36-37). As described above, positivists believe that only objective findings can be classified as true knowledge, and that scientific findings are inherently true (Guba & Lincoln 2005, p.195; Crotty 1998, pp.26-29); science does not ascribe meanings, it "discovers meaning" (Crotty 1998, p.27). Similarly, post-positivists also believe in objectivism, although it is their view that scientific knowledge can never be entirely objective (Guba & Lincoln 2005, pp.193-196; Crotty 1998, pp.26-34); findings are not true, but rather "probably true" (Guba & Lincoln 2005, p.195). In post-positivism, validity is provided by the acceptance of the research by peers with a 'critical eye' to its quality (Guba & Lincoln 2005, p.195; Creswell 2013, p.36). In contrast to the objectivism of positivism and post-positivism, constructivists/interpretivists believe that findings are subjective, and are jointly created by researchers and participants (Guba & Lincoln 2005, p.195; Creswell 2013, p.36); meaning does not exist without consciousness and is "not discovered but constructed" (Crotty 1998, pp.42-43). Constructivists/interpretivists will "rely as much as possible" on the views of participants (Creswell 2013, pp. 24-25). Critical theorists also believe that findings are subjective (Guba & Lincoln 2005, p.195). They believe that "reality is known

through the study of social structures, freedom and oppression, power, and control", and that research can change reality (Creswell 2013, p.37). In critical theory, findings are mediated by values (Guba & Lincoln 2005, p.195).

Axiological assumptions relate to "the role of values" (Creswell 2013, p.21). In both positivism and post-positivism values and biases should be controlled and kept separate to the research, and knowledge is valuable for being knowledge (Creswell 2013, p.36; Crotty 1998, p.27; Guba & Lincoln 2005, p.198). However, in constructivism/interpretivism and critical theory values are considered to be a part of the research. In constructivism/interpretivism the values of those involved with the research are considered to be important and "are negotiated among individuals". In critical theory there is an emphasis on the diverse nature of values within communities (Creswell 2013, pp.36-37). In both paradigms knowledge has value "as a means to social emancipation" (Guba & Lincoln 2005, p.198).

In terms of methodological assumptions and as described above, positivists believe that methodology should involve experimentation and manipulation, and aim to verify hypotheses. Positivists will normally use quantitative methods (Guba & Lincoln 2005, p.195). Constructivists/interpretivists are more likely to use qualitative methods, with a literary style of writing and an inductive, emergent approach to research (Robson 2002, pp.25-27; Creswell 2013, p.36). In contrast, critical theorists begin their research with assumptions about societal, identity or power struggles, document these struggles and use their research to "call for action and change" (Creswell 2013, p.37). Post-positivists are similar to positivists in that they use experimentation and manipulation. However, as described above, post-positivists aim to disprove rather than verify hypotheses. Research in this paradigm may include both quantitative and qualitative methods and will normally follow a deductive, rather than inductive, approach (Guba & Lincoln 2005, p.195; Crotty 1998, pp.26-34; Creswell 2013, p.36).

4.1.1.4 Justification and Pragmatism

This research followed a post-positivist paradigm for two main reasons. Firstly, the assumptions of post-positivism are those to which the author most closely agreed, particularly in relation to ontology and axiology. Secondly, the paradigm allowed the research questions to be thoroughly and appropriately explored. For example, and as described below, the research involved both quantitative and qualitative research. If a positivist rather than post-positivist paradigm had been used, this may not have been appropriate. Another example relates to values. Due to the current and contentious nature of the topic of research, it was considered important for values to be

excluded from most of the research (with the exception of some interview and open-ended survey questions). This fits with a post-positivist paradigm, but would sit uneasily with others, such as constructivism/interpretivism or critical theory, where values play a more central role.

Although a post-positivistic paradigm has been followed in the majority of the research, in some parts this has not been possible. For example, the survey population was chosen via a convenience-sampling method (see section 4.3.2.2), which is contrary to the emphasis on reliability, validity and rigour of post-positivism. The interviews contained some questions that were analysed in an emergent, inductive way, and the content analysis was necessarily subjective. However, if these parts of the research had been avoided due to their violation of post-positivistic principles it would have severely constrained the research. Therefore, in these instances post-positivism was tempered by the pragmatist paradigm, which is focused on research outcomes rather than methods, and allows researchers to choose the most appropriate method in a given situation (Creswell 2013, pp.28-29).

4.1.2 Research Design

Research design can be broadly split into three different approaches: quantitative, qualitative and mixed methods (Creswell 2009, p.3).

4.1.2.1 Quantitative

Quantitative research involves statistically analysing numerical data in order to examine relationships between variables. The purpose of quantitative research is to test "objective theories" (Creswell 2009, p.4) via a "deductive approach" (Bryman 2008, p.22). Methods commonly used in quantitative research include experiments and surveys (Creswell 2009, p.17). A quantitative approach is useful in research that requires measurement, aims to explain causality, and in which it is desirable that results are generalizable and replicable (Bryman 2008, pp.155-157). Criticisms of quantitative research include that it ignores the individual's interpretive ability and treats them the same as inanimate objects, that survey instruments and similar measures are not as accurate and precise as researchers assume, that "the connection between research and everyday life" is hindered by "the reliance on instruments and procedures" and that an objective analysis of variables' relationships does not match the meaning and interpretation of everyday life (Bryman 2008, pp.159-160).

4.1.2.2 Qualitative

Unlike quantitative research, qualitative research is focused on words rather than numerical data (Bryman 2008, p. 22), and aims to understand and explore the meanings that "individuals or groups ascribe to a social or human problem" (Creswell 2009, p.4). Qualitative research is normally analysed inductively, with the aim of generating, rather than testing, theories (Bryman 2008, p.22). Methods commonly used in qualitative research include interviews, open-ended survey questions, observations and the analysis of document, audio-visual, textual and image data (Creswell 2009, p.15). A qualitative approach is useful in research where the aim is to see "through the eyes of" participants, where description, context and process are considered important, where flexibility is important and where there is a desire for "theory grounded in data" (Bryman 2008, pp.385-390). Criticisms of qualitative research include its subjectivity, the difficultly of a qualitative study to replicate, the difficulty of generalizing the research and the common "lack of transparency" in research reports (Bryman 2008, pp.391-392).

4.1.2.3 Mixed Methods

This research can be classified as using a mixed-methods approach. Research employing a mixed methods approach utilizes both quantitative and qualitative research methods (Bryman 2008, p.603). Although some argue that, since quantitative and qualitative methods are rooted in different "epistemological and ontological commitments", a mixed-methods approach is undesirable (Bryman 2008, p.604), the approach actually has a long history of use in research, if one that has often received less attention than quantitative and qualitative approaches alone (Tashakori & Teddlie 2003, pp. 3-8). The mixed-methods approach aims to combine quantitative and qualitative methods in a way that creates research that is stronger than either approach could manage individually (Creswell 2009, p.4). It allows both "confirmatory and exploratory questions" to be answered at the same time (Tashakkori & Teddlie 2003, p.15), whilst minimizing or neutralizing the biases and limitations of quantitative and qualitative approaches when used alone (Creswell 2009, p.14).

There are many different reasons for using a mixed-methods approach. In some research the goal is triangulation, in which the results of both approaches are compared to check that they reach the same conclusion. In other research, the use of mixed methods is considered to increase the credibility of the results. A mixed-methods approach has been used in this research for completeness i.e. it allows a more complete picture of the research area to be created (Bryman 2008, pp.609, 612). Various parts of the research are much more suited to either quantitative or

qualitative methods. For example, the assessment of the effect of DTC genetic tests on health behaviour was stronger for the use of a quantitative approach. However, the in-depth investigation into how and why the information from a test can cause a change in health behaviour required the use of qualitative interviews (Creswell 2009, pp.15-18). Choosing either a solely quantitative or qualitative approach would have substantially limited the research possibilities.

The mixed-methods research design employed in this research was broadly similar to Creswell and Plano Clark's Explanatory Sequential Design (2011, pp.69-71). In Explanatory Sequential Design, the research begins with a quantitative study. Any interesting or significant results are then followed up by a qualitative study, which explores them in more detail. This has the advantage of allowing both an assessment of relationships and trends and an investigation into their causes. This research is similar to that approach, but slightly different; the first quantitative study (the survey) was followed up by both a qualitative study (email interviews) and another quantitative study (a content analysis). This design is shown in Figure 9. As can be seen in the figure, each study had inputs from the research questions and the literature, both the email interviews and the content analysis also had inputs from the survey.

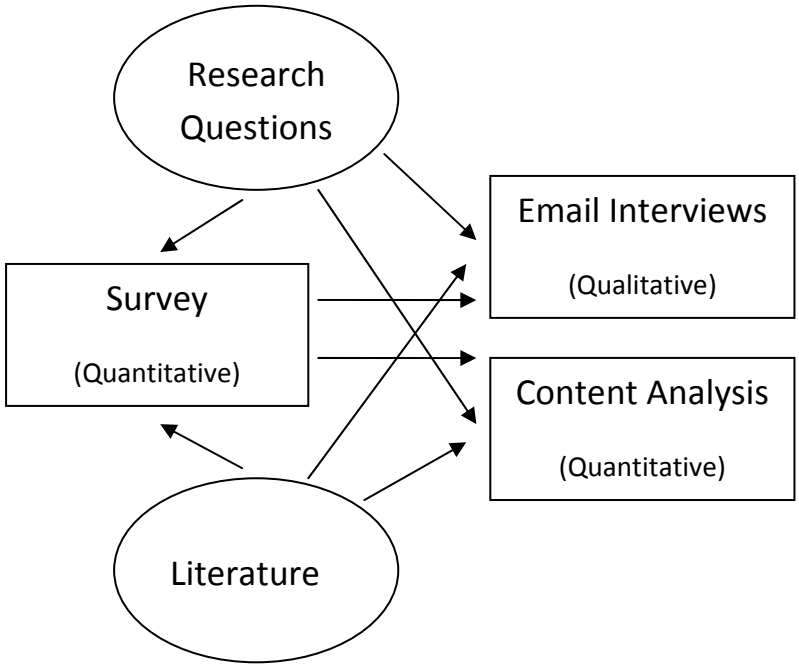


Figure 9 Research design including all three studies and their inputs.

4.2 Previous Studies

Before describing the methods used in this research, it is useful to give a brief description of the methods used in previous studies in this area.

As stated previously (section 2.5), very few studies have examined the effects of DTC genetic tests on health behaviour and/or health anxiety. Gordon et al (2012) collected data through use of semi-structured interviews. Sixty participants were interviewed, all of whom had been provided with a free test that analysed genetic risks for eight different diseases. Kaufman et al (2012) conducted a large-scale survey of 1048 individuals. Respondents were 'real' consumers of genetic tests, and were contacted through three providers of DTC genetic tests. Bloss et al provided 2037 participants with a subsidised DTC genetic test. Symptoms of health anxiety, intake of fat and level of exercise were all measured before receipt of test results, after three months and after a year. Wasson et al provided 20 participants with a free DTC genetic test and interviewed them four times over the course of a year.

As described in section 2.5, the only studies to have analysed the informational aspects of the tests have been content analyses. Relevant studies are: Lachance et al (2010), Geransar and Einsiedel (2008), Einsieldel and Geransar (2009), Hennen et al (2010), Singleton et al (2012) and Liu and Pearson (2008, pp.135, 138-139). Each study identified all of the relevant providers of DTC genetic tests and compared the information on their websites with a set of criteria. The difference between the studies is the formulation of these criteria. Lachance et al (2010) used two sets of criteria in their content analysis. The first set was used to assess the quality and quantity of information on the websites; this was formulated from the recommendations that various professional associations have made as to the information that should be provided for consumers. The second set of criteria was for the usability of the websites and information; this was based on the guidelines published by experts in health literacy. The study by Hennen et al (2010, p.172) was similar, but with only one set of criteria, formulated in this case from a combination of "professional genetic counselling standards" and the literature to create twelve information topics. Topics were assessed as either covered or not, with no assessment of the quality or quantity of information provided. In the study by Liu and Pearson (2008), the criteria was based on a combination of previous research in the area and research into the marketing of prescription drugs to consumers. Geransar and Einsiedel (2008) created their criteria by coding all of the information on providers' websites that related to four different areas. In their second study, Einsiedel and Geransar (2009) again used providers' websites to create their criteria, however this time they created two sets. One set was based on the coding of all information on the websites relating to one specific area; the other by imagining the questions which consumers would ask when searching for information on the websites. Finally, Singleton et al (2012) developed their criteria for the analysis themselves, basing it on the three areas (risks, benefits and limitations) that they wished to examine. They also included any further items which emerged during the analysis.

4.3 Survey

4.3.1 Background

In order to answer the first research question *What effect does the information from a DTC genetic test have on consumers' health behaviour and health anxiety?*, and begin to answer the others, the first study was chosen to be a survey of consumers of DTC genetic tests. As well as actual consumers of the tests, individuals who were strongly considering purchasing a test but had yet to do so were also included in the study, and were designated potential consumers. This inclusion allowed a comparison between two similar groups who differed in receipt of genetic test results.

It was decided that the survey would take the form of a questionnaire. This was due to the large amount of data it was necessary to collect about a variety of topics, including participants': information needs, information-searching behaviour, health behaviours, health anxiety and demographics. Although interviews, for example, would have provided deeper and richer data than a questionnaire, they are much more time-consuming, and so unsuitable for large samples (Gillham 2000, pp.9-11); as a sample size sufficiently large to test for statistical significance was needed, a questionnaire was considered to be more suitable. Also, part of the survey involved questions about health risks and health behaviours, and many participants are more comfortable answering these types of questions with the anonymity of a questionnaire (Czaja & Blair 1995, p.35).

As causation can only be established by use of an experiment where a variable is manipulated (Brace et al 2006, p.94), the survey did have the disadvantage of an inability to prove causation. However, the alternative, providing a large sample of participants with a DTC genetic test and monitoring their health behaviour and health anxiety, was far too expensive to be practical. Even if this alternative study had been possible, the large possibility for bias would have been an issue, as participants who had been offered a free test may have had very different responses to 'real users' i.e. early adopters who had spent a lot of money purchasing a test themselves.

4.3.2 Data Collection

4.3.2.1 Questionnaire Design

4.3.2.1.1 Design Process

It is obviously important that a questionnaire is properly designed and administrated. As Frazer and Lawley (2002, p.2) put it: "a well-designed and administered questionnaire can provide the data necessary to address research questions while a poorly designed and administered questionnaire will result in useless information".

The questionnaire was intended for two separate groups. The first group consisted of people who had purchased a DTC genetic test (consumers) and the second group of people who were considering doing so (potential consumers). Because of the difficulty of designing one questionnaire to cover both groups, two questionnaires were created, with participants clearly directed to the correct one. The questionnaires were hosted on Bristol Online Surveys.

During the design process, special care was taken that the wording of questions was carefully thought through so that they were easy to understand, unbiased and concise, that the questions would provide suitable data for planned analyses, that the questions would provide adequate information for answering the research questions and that no irrelevant questions were asked (Fink 2005, pp.4, 6, 13, 18-19).

A copy of the questionnaires can be found in Appendix A. It should be noted that despite numerous readings, iterations and piloting (as described below) two spelling mistakes remained in the final version of the questionnaires: in question 26 vigorous was wrongly spelt as vigerous and receiving wrongly spelt as recieving.

4.3.2.1.2 Question Explanations

Although most of the questions are self-explanatory with regard to the information that they were designed to collect, there are a few explanations and points that should be mentioned.

One of the important demographic variables measured was socioeconomic status. The method chosen to measure this was the National Statistics Socio-economic Classification (NS-SEC). The NS-SEC is an "occupationally based" method provided by the Office for National Statistics and is "available for use in all official statistics and surveys". The NS-SEC can be used to divide socioeconomic status into eight groups, five groups or three groups depending on the data available. A "self-coded method" is provided, which contains questions specifically designed for questionnaires and produces five separate groups. This method was chosen as the most appropriate for questionnaires, and a sixth group was added to include students (Office for National Statistics [n.d.]).

In order to investigate the effect of the information from the tests on health behaviours, two sets of questions were asked. The first set simply asked consumers if their health behaviour had changed at all since receiving their results, and if so how it had changed. Although this generated important information, there would have been three problems to just using this method and no other to assess changes to health behaviour. Firstly, it is difficult to use the answers to this set of questions to estimate the size of any effect on health behaviour. Secondly, respondents may not have known whether the test had or had not affected their health behaviour; any effect may have been

unconscious, the test may have been one of many factors that affected behaviour or it may have been purchased too long ago to remember accurately. Thirdly, there is always a potential for bias in this type of question, especially considering the high price paid for most of the tests. Therefore, the second set of questions sought directly to compare the health behaviours of consumers and potential consumers, to see if there were any significant differences between the two groups. In order to achieve this it was necessary to probe deeply into respondents' health behaviour by asking detailed questions about specific behaviours. In total, questions about seven different health behaviours were included in the questionnaire: level of fat in diet, level of fibre in diet, level of salt in diet, whether five portions of fruit and vegetables were eaten per day, amount and frequency of alcohol consumption, whether the participant smoked and amount of exercise per week. These particular behaviours were chosen as they are those on which health recommendations commonly focus, with the NHS Choices website used as inspiration (NHS Choices [n.d.]). Health recommendations were used as the basis for these questions for two main reasons. Firstly, it was considered that if an individual was seeking to improve their health behaviour then it is likely that they would focus on behaviours for which it is commonly recommended for people to aim. As a comprehensive list of health behaviours was precluded due to a desire to keep the survey to a reasonable length in order to maximise response rate, it was felt that a focus on these behaviours would therefore increase the likelihood of identification of changes to health behaviour. Secondly, it was considered that recommended health behaviours would be those which are most effective for improving health, and so therefore those on which it would be most useful to focus. However, it should be noted that one limitation of this approach was the use of UK health recommendations for an international study in which respondents reported residency in 22 different countries and in which by far the largest group were resident in the USA i.e. any differences in health behaviour recommendations between different countries may have affected the results. The format of the questions was inspired by a comprehensive health behaviour survey published by the UCL Research Department of Epidemiology and Public Health (UCL [n.d.]), with an aim of creating questions detailed enough to assess whether or not respondents regularly followed the individual health behaviours. Inspiration for the exercise-based questions was also provided by the NHS Choices website (NHS Choices 2011) which provided examples of exercise at different levels of intensity which allowed the questions to be designed to identify how vigorously respondents regularly exercised as well as the length of time for which they did so. As with health behaviour, and for the same reasons, two sets of questions were used to assess any effect of the tests on health anxiety: respondents were first asked if their health anxiety had changed, and if so, how. They were then asked about their current levels of health anxiety, which were compared between the two groups.

The questions on diet asked how respondents' diet compared to recommended levels for salt, fat and fibre intake. This gave rise to a potential error, as respondents may not have known the recommended levels (although a 'don't know' option was included). Unfortunately, it proved impractical to specify recommended levels, as the questionnaire was aimed at an international population, and many countries have different recommended levels. However, this was considered to be a minor source of error; the important information needed for this research was not necessarily respondents' actual health behaviours (although that was the ultimate goal), but their perception of their health behaviours. If an individual believed their salt intake to be no higher than recommended levels, it did not really matter for the analysis if they were mistaken and their salt level was actually above recommended levels; it is the intention of healthy behaviour that was important (any differences between believed and actual health behaviours is then simply a matter of education).

Question 14 (of the consumers' questionnaire, question nine of the potential consumers' questionnaire) was included in order to investigate the ways in which respondents searched for information about DTC genetic tests. Due to the paucity of research in this area the question was designed to be exploratory and open-ended and as such it was considered essential to avoid any influence on respondents' answers. Therefore the question included no prompts or examples other than to include what sources were used, what information was searched for and how successful the search was, and the question was kept as a single, general question rather than split into several questions focused on different aspects. It should be noted that a potential limitation of this approach was that respondents may have been unsure about what information to include and so may have given shorter answers containing less information than could have been gained through a different style of questioning. However, it was considered that this limitation was worth accepting in order to keep potential influence to a minimum.

4.3.2.1.3 Piloting

Two draft questionnaires were created, one for consumers and one for potential consumers. These were modified through various iterations until they were deemed ready for piloting. The piloting was carried out with PhD students, mainly fellow students from the Department of Information Science (with several students from different departments to add to the breadth of knowledge). Those participating in the pilot completed a questionnaire (they were asked to pretend that they had either purchased or were thinking of purchasing a test) and recorded any suggestions at the end. Although ideally those piloting a questionnaire would be members of the population who would be completing the actual questionnaire, the expected difficulty of contacting a large number

of potential respondents precluded their participation in the pilot. The main focus of the pilot was therefore on the clarity of the questions, for which no particular knowledge of DTC genetic tests was needed. Participants' answers were examined for any misunderstandings, their suggestions were considered and any relevant modifications were made. A list of their suggestions can be found in Appendix B.

4.3.2.2 Gathering Responses

Potential participants were contacted through social media; the vast majority through Twitter (by contacting people who 'follow' one of the providers of DTC genetic tests). A convenience-sampling method was used. Unfortunately, convenience sampling can create a selection bias; those who were contacted about the questionnaire may have differed in important ways from those who were not. For example, the method of contacting participants excluded people who were not computer literate or who did not use social media. Also, with convenience sampling it is impossible to know the chance of a random member of the study population receiving an invitation to complete the questionnaire (Fink 2005, pp.45-57 and Czaja & Blair 1995, pp.107-113). However, with such a small study population (only a very small percentage of people have so far purchased genetic tests), it was the only feasible way of contacting participants that retained independence from the companies that sell the tests. As one needs to 'follow' someone on Twitter in order to send them a message, and 'following' a very large number of people without them 'following' you would be considered SPAM by Twitter (Twitter help centre 2011), it was not possible to send a message to everyone who 'followed' a particular company. Instead, it was necessary to search through each user's profile for a link to a blog, AboutMe page or other website. Although not every user had a link to one of these in their profile, a small proportion did. These other websites could then be searched for an email address, or a message could be left in a blog. The response rate was low, estimated at approximately one in five.

Potential respondents were asked to click on a link that took them to a participant information webpage. This page briefly described the research and details about participating (such as the right at any point to withdraw their results) as well as contact details, a link to a page with further information and a link to each questionnaire. A copy of this page and the 'further information' page can be found in Appendix C.

4.3.3 Data Analysis

Most of the methods of analysis are briefly described in appropriate places in the Results chapter. However, there are some parts of the data analysis that require a full explanation.

4.3.3.1 Genealogy-related Purchases

A question was included in the survey to investigate respondents' main reasons for purchasing a DTC genetic test. Many of the tests contain information about ancestry as well as health, and a total of 50 participants only gave genealogy as their answer. This does not affect the analysis; they were still exposed to the health information in their results and helped to ensure that the study sample was representative of the population of people who purchased the tests. Nevertheless, some of the analyses were performed twice, once with the complete results and once with those who only gave genealogy as their answer deleted, to see if this affected the results. It is clearly stated in the results where this occurred.

4.3.3.2 Consumers who had not yet received their results

A small proportion of those who answered the consumers' questionnaire were individuals who had purchased a test but not yet received their results. These individuals were included in the consumers group for most of the applicable results. However, for the comparison of consumers' and potential consumers' health behaviour and anxiety, they were instead included in the potential consumers group. This was because, in order to assess the effects of the test results on health behaviour and anxiety, a comparison was needed between those who had received test results and those who had not, rather than between those who had purchased a test and those who had not. To ensure that potential consumers and consumers who had not yet received their results were not significantly different they were compared with a two sample t-test.

4.3.3.3 Normality and Parametric versus Non-Parametric Tests

For the majority of the quantitative analyses, the data was tested for normality to determine whether a parametric or non-parametric test for significance should be used. Normality was checked in two ways. Firstly, the values for skewness, standard error of skewness, kurtosis and standard error of kurtosis were found. The value of skewness was divided by the standard error of skewness and the value of kurtosis was divided by the standard error of kurtosis. If the resulting values were in the range of -1.98 to 1.98, the distribution was considered approximately normal; if outside the range they were considered non-normal. Secondly, a one-sample Kolmogorov-Smirnov test was conducted to compare the distribution with a normal distribution; if the p value was over 0.05 it was considered normally distributed and if under it was considered non-normally distributed.

If the data were found to be non-normal, three types of transformation were attempted to attain normality: a square-root transformation, log transformations and an inverse transformation (Osborne 2002). On no occasion did these transformations actually produce normality.

If the data were non-normal, a non-parametric test of significance was used instead of a parametric one. For situations where the parametric choice would be an independent t-test (i.e. comparing two groups) the non-parametric alternative would normally be the Mann-Whitney U test (Brace et al 2006, p.85). However, most of the data contained a large number of ties, such as the health-behaviour scores and the Likert scores, and the Mann-Whitney U test is only suitable if there are a small number of ties (Rice 2007, p.437). Therefore in these situations a two-sample Kolmogorov-Smirnov test was used instead.

4.3.3.4 Difference in Health Behaviours between Consumers and Potential Consumers

The questionnaire contained seven questions on health behaviours. However, the responses for the question on alcohol consumption were too incomplete for use in the analysis as many respondents did not accurately specify the quantity they normally drank; therefore only the other six were used.

Each respondent was assigned a score of one or zero for each question depending on whether they were following health recommendations or not. For the questions on salt and fat consumption, those who ate lower than or equal to recommended amounts scored one, whilst those who ate higher than recommended amounts scored zero. For fibre, those who ate higher or equal to recommended amounts scored one, whilst those who ate lower than recommended amounts scored zero. Those who ate five fruit and vegetables every day or most days scored one, whilst those who ate them on some days or less scored zero. Those who never smoked cigarettes, cigars or pipes, or who only smoked less than one cigarette per day and/or cigars or pipes very occasionally, scored a one, whilst those who smoked more often scored zero. For exercise, those who did 150 minutes moderate exercise or 75 minutes vigorous exercise a week (or a combination of the two) (NHS Choices 2011) scored one, whilst those who did less scored zero.

These scores were combined to give the respondents a health-behaviour score from zero to six. The health-behaviour scores of the consumers and potential consumers groups were then compared.

4.3.3.5 Website-Assessment Statements

One of the questions concerned respondents' assessment of the website of the company from which they purchased their test, or the company from which they were thinking of purchasing a test. Respondents were asked to rank how much they agreed with six statements on a Likert scale. This

question was originally designed to compare different companies' websites. Unfortunately, most respondents purchased their test (or were thinking of purchasing a test) from 23andMe, so this analysis became impossible. However, the questions did provide some data useful for other analyses. They are referred to in the results as website-assessment scores.

For one analysis, a mean was taken of each respondent's combined scores for the different statements. This mean score is referred to in the analysis as the mean website-assessment score. The sixth statement, *I had to look at other sources to find enough information to make a decision about buying a test*, was excluded, as it was too different from the other statements; it would therefore be much more influenced by the information-seeking behaviour of the respondent than by the information content of the website alone.

4.3.3.6 Correlation

Several parts of the analysis involved an assessment of whether or not there was a significant correlation between two variables. In each instance a two-tailed Spearman's Rank correlation coefficient was calculated. It should be noted that this use of a two-tailed test is a potential limitation of the analysis as it generates the possibility of false negative results, especially with the comparison of variables such as perceived risk and health behaviour where there is a reasonable reason to believe that a correlation might exist. However, the decision to use a two-tailed test in each instance was taken due to the paucity of research in the area; it was considered dangerous to make assumptions about the relationship between variables and, due to the importance to the field of any significant findings, better to risk false negative than false positive results.

4.3.3.7 Weighting

In order to determine if the significant difference between consumers and potential consumers in health-behaviour scores was due to underlying demographic variables, a two-sided Fisher's Exact test was used to identify any significant differences in the proportion of the consumers and potential consumers in each demographic included in the survey (only respondents who had answered enough questions to be given a health-behaviour score were included). A Fisher's Exact test was used instead of a Pearson's Chi-Square test due to the small cell size for some of the categories. For three of the demographic variables (socioeconomic status, ethnicity and country of residence), a large majority of respondents were in one category, with a corresponding small *n* in the other categories. Therefore the categories for these demographics were collapsed, to compare the managerial and professional occupations category against the other categories combined (for

socioeconomic status), to compare Caucasian ethnicity against other ethnicities combined (for ethnicity) and to compare residency in USA against residency in all other countries combined (for country of residence).

The health-behaviour scores were then weighted to control for the different demographic variables and compared with a two-sample Kolmogorov-Smirnov test. As above, the categories for socioeconomic status, ethnicity and country of residence were collapsed, due to the large number of respondents in one category in each of the three demographic variables.

A stepwise multiple regression was then performed in an attempt to build a model of all predictor variables with health behaviour as the criterion variable. As above, the categories for socioeconomic status, ethnicity and country of residence were collapsed, due to the large number of respondents in one category in each of the three demographic variables.

4.3.4 Comparison with Previous Studies

The design of this study allowed a unique viewpoint when compared with the published studies in this area. Bloss et al (2010, 2011b, 2013) and Gordon et al (2012) provided participants with a heavily-subsidised or free test. As described above, this may have created results which were not able to be generalized to 'real users' of DTC genetic tests. Although Kaufman et al (2012) was a survey of 'real users' of the tests, the nature of the study was such that the results were entirely reliant on respondents' correct and unbiased recall of changes to their health behaviour. Although part of this study was similarly limited, the bias has been mitigated by the inclusion of a direct comparison of consumers' and potential consumers' current health behaviour and health anxiety; thus providing results that were based on 'real users' but did not entirely rely on their correct recall.

4.4 Email Interviews

4.4.1 Background

The survey fully answered the first research question, and began to answer the others. In order to address the second, third, fourth and sixth questions:

- How does the information from a DTC genetic test effect consumers' health behaviour and health anxiety?
- What are consumers' information needs and information-seeking behaviours?
- What effect does the information from a DTC genetic test have on consumers' information needs and information-seeking behaviours?

- What are consumers' opinions about, and experiences with, DTC genetic tests?

and to begin to answer the fifth question, it was decided that the second study would be email interviews.

Interviews were chosen because they can collect data that is often not accessible with the use of other techniques (Blaxter et al 2001, p.172). They also have the benefit of flexibility (Bryman 2008, p.436). Emails were chosen as a convenient method of communication given the international nature of the study. They also had the advantage that, since some of the questions asked about events and situations that had occurred some time ago, they gave participants time to think about their answers. Although a pre-interview briefing can give interviewees this opportunity in a face-to-face or telephone interview, the use of emails allowed them to think about each question individually.

4.4.2 Study Design

4.4.2.1 Contacting Respondents

In total, 36 respondents participated in the email interviews. Potential interviewees were chosen from those who had answered the consumers' survey, had received their results, indicated that they would be happy to be contacted about further research and given an email address.

Potential respondents were divided into three groups based on their answers to the survey: those whose health behaviour had changed after receiving their results, those whose health anxiety had changed after receiving their results and those who had had no change in either health behaviour or health anxiety (there was some overlap between the first two groups). The third group was then subdivided into those who had used zero or one source to search for information about the tests, those who had used two or three sources and those who had used four or over.

Originally, an aim was set to contact 25 respondents. To achieve this, a random-number generator was used to select five potential respondents from each of the five groups described in the previous paragraph. Each potential respondent was emailed to ask if they would be willing to participate in the interviews. The emails explained the research, gave basic information and a link to a website with further information (see Appendix D). If any respondent did not wish to participate the random-number generator was used to select another potential respondent from their group, until there were five respondents from each group who were willing to participate.

The remainder of those whose behaviour and/or anxiety had changed, and several more whose behaviour or anxiety had not changed, were then contacted; this brought the total to 36. These extra respondents were contacted with the intention of asking about their health behaviour and anxiety, and so only those topics were included in their interviews.

If respondents stopped replying to emails before the interview was finished then reminders were sent, limited to two consecutive reminders per participant.

4.4.2.2 Interview Design

The interviews were semi-structured. Every interview covered a pre-formulated list of topics and issues. Each topic was asked about in a standard way, although an element of flexibility in timing and wording was allowed if deemed appropriate. Any interesting points raised during the interview, either in answer to the questions or raised by the interviewees, were fully followed up with extra questions (Bryman 2008, pp.438-439). Interviewees were also asked if they had anything to add that had not been included in the interview. The interviews followed this semi-structured design as it combined the benefits of structured and unstructured interviews; the structured parts allowed for the collection of "precise data" whilst the flexibility ensured that data was not limited by "a priori categorization" (Fontana and Frey 2000 p.653).

The number of questions per email was limited to one or two. If any clarification or expansion of an interviewee's answer was needed further questions were asked. As stated above, a list of topics and issues was formulated before the interviews began. These were:

- Health behaviour, including change or lack of change due to results and reason for change or lack of change
- Health anxiety, including change or lack of change due to results and reason for change or lack of change
- Reasons for purchasing a test
- Ease of understanding of results
- Any surprise caused by results
- Information need, including responsibility for consumers to have adequate knowledge and rating of information provided
- Information-seeking behaviour, including information searched for and sources used
- The sharing of results with health professionals, including whether or not results had been shared and opinions about sharing
- Basic genetic knowledge

A standard set of questions was developed for each topic and issue. These questions were developed during the interviews, and were flexible if appropriate, and so all of the interviewees did not receive an identical question. The questions were designed to be unbiased and non-judgemental, and to influence interviewees as little as possible. This was sometimes stated explicitly during the interviews. For example, when asking those whose health behaviour had not changed why it had not changed, the following was stated in brackets "please note, I'm not implying that you should have made changes – I'm asking everyone this question, regardless of whether they made changes or not". This was to ensure that the question did not cause interviewees to feel that they should have made changes to their health behaviour.

A good example of a set of questions is those that were asked for the 'sharing results with health professionals' topic. This topic had two standard questions. These were:

Have you shared any of your health results with your doctor or another healthcare professional? If so what was their attitude towards them?

What is your opinion about people sharing their results with their doctors?

However, based on interviewees' answers, more questions were commonly asked after each individual question in order to fully explore the topic.

4.4.3 Analysis

The interview transcripts were analysed via a process of coding. Creswell (2009, p.186) describes coding as "the process of organizing the material into chunks or segments of text before bringing meaning to the information". This approach allows a possibly overwhelming quantity of data to be handled in a manageable way (Robson 2002, p.477). The transcripts were coded using the software package NVivo.

The transcripts were first coded by topic, originally into large general groupings (i.e. behaviour, anxiety, information, other), and then subdivided until appropriately-sized topic nodes were created. These topic nodes were predetermined i.e. based on the list of topics formulated before the interviews began (Creswell, 2009, p.187). The manuscripts were then analytically coded, mainly within the topic nodes but also across them, in order to recognize emerging themes. The codes used in this part of the analysis were those that emerged from the transcripts; they were not predetermined (Creswell, 2009 p.187). The manuscripts were coded and recoded iteratively until it was considered that all of the themes had emerged and their meanings had been properly understood (Richards 2009 pp. 96-97, 102-106).

The findings from the email interviews (along with some from the survey) were modelled in the final discussion to create a model of the purchase of a DTC genetic test and a model of the information-seeking behaviour of consumers of DTC genetic tests (see section 8.6). These models were based on ideas that emerged from the data rather than the testing of predetermined ideas.

4.4.4 Comparison with Previous Studies

Apart from this research, the only studies identified in the literature review that used interviews to investigate DTC genetic tests are Gordon et al (2012) and Wasson et al (2013). Compared with those studies, this research had the advantage (as with the survey) of the inclusion of 'real' consumers of DTC genetic tests, rather than participants who had been provided with a free test. The results from this study may therefore be generalizable in a way that the results from the previous studies are not. Also, due to their small sample size, Wasson et al (2013) only interviewed a few people who had noticed a behavioural or psychological effect. This negatively compares with this research where the large population of survey respondents allowed a reasonable number of such participants to be interviewed.

The interview method of this study allowed for an in-depth examination of the self-reported effects of DTC genetic tests on health behaviour and anxiety, as well as mechanisms by which they occurred. The other two studies in this area, aside from Gordon et al (2012) and Wasson et al (2013), lacked the capability to do this due to the limitations of questionnaires in the study by Kaufman et al (2012) and the lack of participant questioning in the study by Bloss et al (2011b).

In terms of the informational aspects of the tests, all relevant previous studies were content analyses. This study allowed for an assessment of the informational aspects from the consumers' viewpoint, which would not have been possible in the previous studies.

4.5 Content Analysis

4.5.1 Background

The survey and email interviews provided the data to fully answer all of the research questions apart from the fifth: are consumers' information needs met by the information provided on the websites of companies that sell DTC genetic tests? This question was only briefly touched upon by the first two studies, and so to fully answer it a content analysis of all of the websites that sold DTC genetic tests was conducted.

Neuendorf (2002, p.1) defines a content analysis as a "systematic, objective, quantitative analysis of message characteristics". A content analysis involves assessing whether a message (be it text, image or any other construct with meaning) fulfils a list of conditions set out a priori in a codebook, and allows the contents of a message to be scientifically analysed (Neuendorf 2002, pp.10-11,50-51; Krippendorff 2004, pp.3, 18).

Although there have been several content analyses of the websites of providers of DTC genetic tests, they have all been from either the researcher's or professionals' points of view i.e. the content-analysis frameworks have been based on the information that the researcher or relevant professionals think should be provided for consumers. Although this is an important viewpoint when assessing the information provided, it excludes consumers from the process; it is likely that consumers have a different opinion on the information that should be provided than professionals or researchers do. Therefore, this content analysis was designed to take both the viewpoint of professionals and the viewpoint of consumers into account, something which has not been the case for any currently-published study.

4.5.2 Research Design

4.5.2.1 Framework Development

4.5.2.1.1 Background

The initial stage of the research design was the development of a framework or codebook for the assessment of the websites. This framework was composed of a large number of different items, and during the analysis each website was assessed as to whether or not it covered each one.

An important element of content analyses is objectivity (Neuendorf 2002, p.11). Although the actual assessment of whether or not an item was covered by a website was necessarily subjective, an attempt was made to reduce the subjectivity of the study as much as possible. One of the key parts of this was the use of items that were based on external factors rather than simply the creation of items that seemed appropriate. It was also considered important, as is stated above, for the consumer's viewpoint to be represented in the framework. However, it was recognised that the consumer's view of the information that should be provided by the DTC websites may have been incomplete, and so the professional's viewpoint was also included. Therefore, the items were based on two different sources: consumers' and professional organizations' opinions of the information that should be provided on the websites of providers of DTC genetic tests.

4.5.2.1.2 Items Generated from Consumers

The items derived from consumers' opinions were based on their answers to the survey questions about information need and information-seeking behaviour. These answers were collated together and distilled into a set of items. For example, one consumer gave the following answer:

How reliable were the tests. What was the repeatability of the results when done using different testing methods by different companies. I have had my Y DNA tested by three companies and all were in agreement on the counts at the same markers even though each company used a different test method.

This was distilled into the following two items:

Reliability of tests

Similarity of results between different companies

Another consumer gave the following answer:

I mainly Googled around the terms, looking at journal articles and wikipedia pages to educate myself on the material described by these companies. I was looking to understand the tests and make a judgement on their reliability and validity. I was somewhat successful, but not satisfied enough to make a decision whether to buy or not buy.

This was distilled into the following two items:

Reliability of tests

Validity of tests

Appendix E shows the generation of these items.

4.5.2.1.3 Items Generated from Professional Organisations

The second set of items, those based on the professional's viewpoint, was generated from professional organisations' recommendations of the information that should be provided on the websites of providers of DTC genetic tests. A general internet and literature search found 12 professional organisations with recommendations for, or opinions on, the information that should be provided. These were:

- The American Society of Human Genetics
- The American College of Clinical Pharmacology
- The European Society of Human Genetics

- The International Society of Nurses in Genetics
- The American College of Medical Genetics
- The American Congress of Obstetricians and Gynaecologists
- The American Society of Clinical Oncology
- The Austrian Bioethics Commission
- The Belgian Advisory Committee on Bioethics
- The National Council of Ethics for the Life Sciences (Portugal)
- The Nuffield Council on Bioethics
- The Human Genetics Commission

As above, the professional organisations' recommendations and opinions were distilled into a list of items.

For example, the American College of Obstetricians and Gynaecologists stated in their publication:

However, patients are not made aware that failure to indicate results of genetic testing in life insurance or disability applications could be considered fraud. In addition, many laboratories have not indicated their policies on what is done with the DNA sample after analysis. To ensure privacy, DNA samples should be destroyed after the requested test is performed. Those overseeing procedures for testing should continue to work to address patient privacy concerns.

Appropriate pretest and posttest counseling should be provided, including a discussion of the risks, benefits, and limitations of the testing.

This was distilled into the following items:

Legal position of declaring results for insurance

Legal position of declaring results for disability applications

Fate of DNA sample

Privacy issues

Risks of testing

Benefits of testing

Limitations of testing

Appendix E shows the generation of these items.

4.5.2.1.4 Creation of Framework and Data Collection

The two lists of items were combined to create a framework for the content analysis. Any items within or between the lists that were very similar were combined into a single item. However, items with a small amount of variability were kept separate, which allowed for a comprehensive and detailed assessment of the websites. Items were reworded where necessary to ensure a standard format throughout the framework and a clear delineation between items.

It was easy to assess whether most items were covered or not by the websites. However, some items were difficult to assess. In these cases detailed notes were made, and the notes and decisions were continuously reassessed and compared between the websites to ensure that a consistent coding approach had been used. The coding erred on the generous side i.e. if there was difficulty in deciding whether or not a website had covered an item the website received the benefit of the doubt.

It should be noted that several items in the content analysis have specific definitions in scientific terminology that differ from their meaning in 'everyday language'. However, since the focus of the content analysis was on provision of information for consumers, it was decided to interpret these items from the consumers' viewpoint i.e. what it was considered a 'lay person' would understand a term to mean. For example, the item 'accuracy' was interpreted as any information that related to how accurate the test was rather than to a specific accuracy value. One limitation that may have arisen from this decision is that some items may not have been interpreted how they were originally intended. For example, if a survey respondent with a scientific background had made use of scientific terms. However, scientific definitions were used for terms that did not have relevant equivalents in 'everyday language', such as sensitivity, which is the proportion of correctly identified true positives, and specificity, which is the proportion of correctly identified true negatives (Altman & Bland 1994, p.1152).

Other items were not immediately obvious as to their intended meanings. In these instances a 'best guess' was used as to what information should be searched for with a focus, as above, on the consumers' viewpoint. For example, 'diagnostic value of the test' was taken to mean any information about whether or not the test would be useful in the diagnoses of disease, 'general accuracy' was taken to mean any general information about the accuracy of the test rather than specific information such as error rates and 'general coverage' as any general information that related to what the test covered rather than specifics such as coverage of particular SNPs. Although the subjectivity and possible inaccuracy of these interpretations was a potential limitation of the

study, it was considered to be more appropriate than to delete these items and therefore not cover a large body of information that consumers were likely to wish to know.

Several items were removed from the combined framework due to their unsuitability. These are listed, alongside the reason for their removal, in Appendix F.

4.5.2.2 Providers of DTC Genetic Tests

A literature and general internet search was conducted in order to find as large a proportion of the providers of DTC genetic tests as possible.

The literature search resulted in the discovery of The Genetics and Public Policy Centre (GPPC) (2011) which published a list of a large number of providers in 2009 and an updated one in August 2011 (although only tests available in the USA were covered). All of the companies on both lists were copied. An internet search was then conducted to find any providers not included on the list, with the search terms those used by the GPPC to develop their list: genetic test; genetic testing; genomic test; DNA test kit; direct-to-consumer genetic tests; direct-to-consumer genomic tests (The Genetics and Public Policy Centre 2011). The first 200 results for each search term were checked for any provider which was not on the GPPC lists. Each provider from both lists and any additional provider discovered in the internet search were checked to see if they were still trading, and if so what type of genetic test they sold. Any provider which sold a test that examined more than one health condition were included in the content analysis. These were:

- 23andme
- decode Genetics
- easyDNA
- GenePlanet
- Inherent Health/Interleuken Genetics, Inc.
- Lumigenix
- Map My Gene
- Test Country
- Viaguard/Accu-metrics
- Navigenics
- Pathway Genomics
- International Biosciences
- Genetic Health

A table showing all of the providers considered for inclusion, along with the assessment process, is in Appendix G.

Any blogs maintained by providers were not included in the content analysis; it was considered unreasonable to assume that consumers would read through a providers' entire history of blog posts. Several websites contained links to different websites. If it was stated on the page that these links should be followed, or that specific information could be found on them, then the linked page was included; the rest of the website, however, was not.

4.5.3 Content Analysis

The data was analysed in two ways: statistically and thematically.

Descriptive statistics were used to compare the websites and the groups of items. The websites were compared to determine which website covered the most number of items in total, and to compare the coverage of the different groups of items between them. The different items were compared within the groups, to determine which items were most covered and which were least, and the groups were compared to see which groups were covered most and which least. This is explained in appropriate places in the Content Analysis chapter.

The items were then arranged into different themes. These themes were examined to see how well their constituent items were covered, and compared to see which themes had the higher coverage. This is also explained in appropriate places in the Content Analysis chapter.

4.5.4 Comparison with Previous Studies

Although there are several published content analyses of the websites of providers of DTC genetic tests (see section 2.4.2.2), this study differs significantly from them in terms of criteria used to assess the information provision. As described in section 4.2, previous content analyses have either formulated criteria based on professional recommendations, the literature, the websites themselves or simply the areas they wished to investigate. Although one set of the items used in this study were generated from professional recommendations, the other set was generated from consumers' information needs as identified in the survey. This is a very important contribution, as it allowed for an assessment of the provision of information that consumers themselves wished to know, rather than simply what others think that they should know.

5 Survey

This chapter presents, analyses and discusses the results of the survey. Data tables for statistical tests are shown in Appendix H.

5.1 Basic Demographics

5.1.1 Results

There were 225 responses to the **consumers'** questionnaire and 67 responses to the **potential consumers'** questionnaire. In total, 14 of the responses to the **consumers'** questionnaire and three of the responses to the **potential consumers'** questionnaire were deleted. Out of those deleted from the **consumers'** questionnaire, six were blank and one blank apart from one incorrectly-completed question. Three violated the instructions given to participants i.e. that participants should have purchased a test which included information about disease risks, and four specifically stated that they had not looked at the health results or had had them turned off. Out of those deleted from the **potential consumers'** questionnaire, two were blank and one had specifically stated that they were not a fan of the tests and had only followed on Twitter out of curiosity (hence not a potential consumer).

Therefore the final sample contained a total of 275 usable responses, composed of 211 usable responses to the **consumers'** questionnaire and 64 usable responses to the **potential consumers'** questionnaire. This is shown in Table 1.

Table 1 Composition of Final Sample

	Consumers' Questionnaire	Potential Consumers' Questionnaire	Total
Total Responses	225	67	292
Deleted Reponses	14	3	17
Final Sample	211	64	275

Out of the usable responses to the **consumers'** questionnaire, 189 were completed by consumers who had received their results and 22 by consumers who had not yet received their results. As described in section 4.3.3.2, consumers who had not yet received their results were included in the consumers group for all applicable analyses. However, for the analysis of the effect of the tests on health behaviour and health anxiety (section 5.3) they were included in the potential consumers group.

Most demographics are shown for three groups: consumers, potential consumers and a combined group of all of the respondents.

Table 2 shows the proportion of each gender within the respondents. Nearly two thirds of the respondents within each group were male.

Table 2 Gender of Respondents

	Consumers Percentage (N)	Potential Consumers Percentage (N)	Combined Percentage (N)
Male	62.1 (126)	65.0 (39)	62.7 (165)
Female	37.9 (77)	35.0 (21)	37.3 (98)
Total	100 (203)	100 (60)	100 (263)

Figure 10 shows the difference in age groups between consumers and potential consumers. Consumers were generally older than potential consumers, with mode age groups of 30-44 and 18-29 respectively. The ages of the combined group is shown in Figure 11, with a mode age group of 30-44.

Figure 10 Percentage of consumers and Potential Consumers in each age group

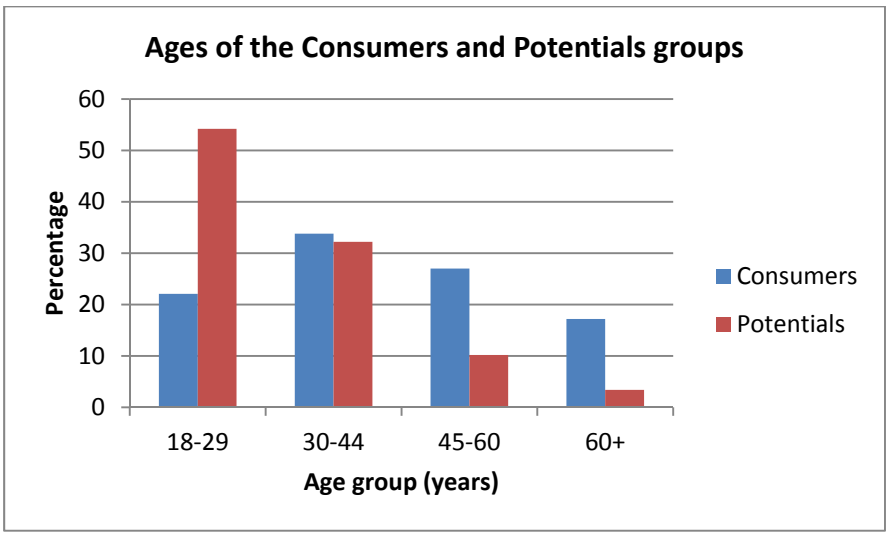
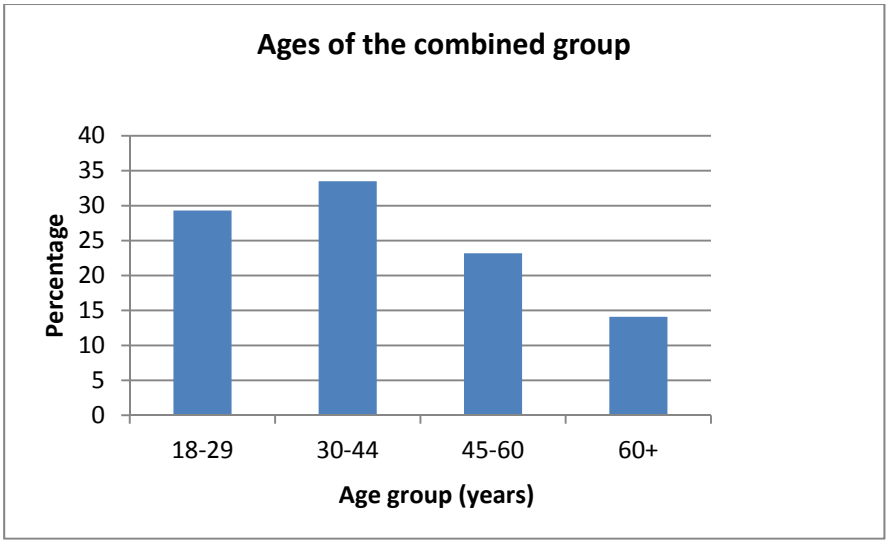


Figure 11 Percentage of Respondents in each age group



The highest level of education reached (but not necessarily completed) amongst the consumers, potential consumers and combined groups is shown in Table 3. The mode level of education for all three groups was a postgraduate degree, followed by a bachelor's degree, with these two levels accounting for over 93% of respondents in each group.

The percentage of respondents in each socioeconomic category is shown in Table 4. As might be expected with early adopters of a new healthcare technology, 83.5% of respondents (85.3% of consumers and 77.6% of potential consumers) were in the highest category: managerial and professional occupations.

Table 3 The education levels of the Consumers, Potential Consumers and Combined groups.

Highest level of Education	Consumers Percentage (N)	Potential Consumers Percentage (N)	Combined Percentage (N)
School	3.5 (7)	3.4 (2)	3.4 (9)
Post-school (e.g. diploma, associate degree etc)	3.0 (6)	3.4 (2)	3.1 (8)
Bachelor's degree	39.1 (79)	42.4 (25)	39.8 (104)
Postgraduate degree	54.5 (110)	50.8 (30)	53.6 (140)
Total	100 (202)	100 (59)	100 (261)

Table 4 The socioeconomic status of the Consumers, Potential Consumers and Combined groups

Socioeconomic category	Consumers Percentage (N)	Potential Consumers Percentage (N)	Combined Percentage (N)
Managerial and professional occupations	85.3 (168)	77.6 (45)	83.5 (213)
Intermediate occupations	4.6 (9)	3.4 (2)	4.3 (11)
Small employers and own account workers	3.0 (6)	6.9 (4)	3.9 (10)
Lower supervisory and technical occupations	1.5 (3)	1.7 (1)	1.6 (4)
Semi-routine and routine occupations	1.0 (2)	1.7 (1)	1.2 (3)
Students	4.6 (9)	8.6 (5)	5.5 (14)
Total	100 (197)	100 (58)	100 (255)

Respondent's ethnicities are shown in Table 5. As they were self-reported ethnicities they have been organised into appropriate categories. Over 84% of respondents (83.8% of consumers and 87.5% of potential consumers) described themselves as Caucasian, White or similar. Ethnicities included in the Other category included Jewish, Hispanic, Mexican, White Latino, African American, Chinese and Iranian.

Table 5 Respondent's Self-Reported Ethnicities

Ethnicity	Consumers Percentage (N)	Potential Consumers Percentage (N)	Combined Percentage (N)
Caucasian	83.8 (165)	87.5 (49)	84.6 (214)
Mixed-race	7.6 (15)	3.6 (2)	6.7 (17)
Asian	3.0 (6)	7.1 (4)	4.0 (10)
Other	5.6 (11)	1.8 (1)	4.7 (12)
Total	100 (197)	100 (56)	100 (253)

Respondents' countries of birth, countries of residence and the percentage who resided in a different country to that of their birth are shown in Tables 6 to 8. Just over two thirds of

respondents (67.2% of consumers and 54.2% of potential consumers) were born in the USA, with an even higher percentage of 70.5% (72.4% of consumers and 63.8% of potential consumers) resident there at the time of the survey. The second most common country of birth and residence was the UK (9.1% and 9.6% respectively) followed by Canada (4.6% and 6.5% respectively). All countries in which only one respondent was born or resident are included in the Other group. For countries of birth this included Argentina, Austria, Bangladesh, Brazil, Bulgaria, Greece, Iran, Ireland, Kuwait, New Zealand, Norway, Pakistan, Poland, Russia, Slovakia, Sweden, Switzerland, Trinidad and Tobago and Uruguay and for countries of residence it included Finland, Argentina, Brazil, Columbia, India, Netherlands, New Zealand, Norway, Spain, Switzerland, Ukraine and Uruguay. Over a fifth of respondents (21.3% of consumers and 24.1% of potential consumers) lived in a different country to that of their birth.

5.1.2 Discussion

Respondents' demographics are broadly similar to those in previous studies of users of DTC genetic tests; namely, participants have been mainly of a white/Caucasian ethnicity, members of a high socioeconomic or income group, possessed a high level of education and have been in a middle age range (Kaufman et al. 2012 & Bloss et al. 2011b). These findings are unsurprising for early adopters of a new and relatively expensive healthcare technology.

One result that is surprising is that nearly two thirds of respondents were male. As participants in the studies conducted by Kaufman and Bloss were nearer to 50% male and female it is not likely that this reflects the consumer population; it is possible that it is due to a bias in the sampling method (see section 4.3.2.2) but if so it is unclear what this would be.

Respondents represented a wide variety of different countries, both in birth and in current residence, but the majority were born or lived in the USA. Again, this is unsurprising, especially given the large proportion of providers of DTC genetic tests that are located in the USA. Respondents were also a highly mobile group, with over a fifth living in a different country to that of their birth.

There was only one significant demographic difference between consumers and potential consumers and that was of age group, with consumers more likely to belong to an older age group. This could be due to numerous reasons. For example, it is possible that the high price of the tests causes them to be unaffordable for some younger potential consumers. Alternatively, there may be a difference in opinions about the tests between individuals of different ages. This is an area that has not been examined in this research and may benefit from further study.

Table 6 Respondents' countries of birth

Country of Birth	Consumers Percentage (N)	Potential Consumers Percentage (N)	Combined Percentage (N)
USA (including born abroad to US parents)	67.2 (137)	54.2 (32)	64.3 (169)
UK	9.3 (19)	8.5 (5)	9.1 (24)
Canada	2.9 (6)	10.2 (6)	4.6 (12)
Australia	3.4 (7)	0.0 (0)	2.7 (7)
Germany	1.5 (3)	3.4 (2)	1.9 (5)
France (including overseas Depts)	2.0 (4)	0.0 (0)	1.5 (4)
Spain	1.0 (2)	1.7 (1)	1.1 (3)
India	0.5 (1)	3.4 (2)	1.1 (3)
Philippines	0.5 (1)	3.4 (2)	1.1 (3)
Finland	1.0 (2)	0.0 (0)	0.8 (2)
Columbia	1.0 (2)	0.0 (0)	0.8 (2)
Denmark	0.5 (1)	1.7 (1)	0.8 (2)
Italy (including Vatican City)	1.0 (2)	0.0 (0)	0.8 (2)
Netherlands	0.5 (1)	1.7 (1)	0.8 (2)
South Africa	0.5 (1)	1.7 (1)	0.8 (2)
Ukraine	0.5 (1)	1.7 (1)	0.8 (2)
Other	6.9 (14)	8.5 (5)	7.2 (19)
Total	100 (204)	100 (59)	100 (263)

Table 7 Respondents' countries of residence

Country of Residence	Consumers Percentage (N)	Potential Consumers Percentage (N)	Combined Percentage (N)
USA	72.4 (147)	63.8 (37)	70.5 (184)
UK	9.4 (19)	10.3 (6)	9.6 (25)
Canada	5.9 (12)	8.6 (5)	6.5 (17)
Australia	3.9 (8)	0.0 (0)	3.1 (8)
Denmark	0.5 (1)	3.4 (2)	1.1 (3)
France (including overseas Depts)	1.5 (3)	0.0 (0)	1.1 (3)
Italy (including Vatican City)	1.5 (3)	0.0 (0)	1.1 (3)
Sweden	0.0 (0)	1.7 (1)	0.8 (2)
Germany	0.5 (1)	1.7 (1)	0.8 (2)
Ireland	0.5 (1)	1.7 (1)	0.8 (2)
Other	3.4 (7)	8.6 (5)	4.6 (12)
Total	100 (203)	100 (58)	100 (261)

Table 8 Percentage of respondents who live in a different country to that of their birth

Live in a different country from that of birth?	Consumers Percentage (N)	Potential Consumers Percentage (N)	Combined Percentage (N)
Yes	21.3 (43)	24.1 (14)	21.9 (57)
No	78.7 (159)	75.9 (44)	78.1 (203)
Total	100 (202)	100 (58)	100 (260)

5.2 How Respondents First Discovered DTC Genetic Tests

5.2.1 Results

One question in the survey asked respondents how they first found out about DTC genetic tests. The results (with consumers and potential consumers combined) are shown in Table nine. Over a third of respondents found out about the tests through blogs, making it the most common source. This was followed by friends, family and other social connections, which was mentioned by just under 30% of respondents. The third highest source was the websites of a company that sells DTC genetic tests, mentioned by just under a quarter of respondents. No total is included in the table as some respondents gave more than one answer.

5.2.2 Discussion

The results show that respondents first heard about DTC genetic tests from a wide variety of different sources. This is an interesting finding and shows the extent to which knowledge of and information about DTC genetic tests is beginning to spread.

One notable finding is that over a third of respondents stated that they heard about DTC genetic tests through blogs. This result may have been influenced by the sampling method used (see see section 4.3.2.2); since respondents were mainly contacted through social media (including blogs) it is likely that this created a bias towards individuals who commonly use such websites. However, the fact that the tests are only available online, that they are a new technology mainly purchased by early adopters, and that there is a large quantity of information about the tests available on blogs may also help to explain it.

Table 9 How respondents found out about DTC genetic tests

Source	Percentage (N)
Blogs	35.7 (97)
Friends, family, colleagues, connections and conversations	29.2 (79)
The website of a company that sells genetic tests	24.4 (66)
Other internet site	17.3 (47)
Google	18.5 (50)
Magazine	13.3 (36)
Twitter	13.3 (36)
Newspaper	8.5 (23)
Advertising	7.4 (20)
Wikipedia	5.5 (15)
Mailing Lists and Forums	5.5 (15)
TV (Other)	4.1 (11)
Work-Related	3.7 (10)
TV (News)	3.0 (8)
Conferences	2.2 (6)
Facebook	1.8 (5)
General Knowledge	1.8 (5)
Doctor	1.5 (4)
Books	1.5 (4)
School, University or Training	1.5 (4)
From Genealogy	0.7 (2)
General Internet Research	0.7 (2)
Miscellaneous Media	0.7 (2)
Previous Genetic Testing	0.7 (2)
Genetics Society	0.4 (1)
Lectures	0.4 (1)
Own Research	0.4 (1)
Radio Interview	0.4 (1)
The Quantified Self Movement	0.4 (1)
Through a Company	0.4 (1)

5.3 Effect of the Information from DTC Genetic Tests on Health Behaviours and Anxiety Levels

5.3.1 Stated Changes in Behaviour and Anxiety

Consumers were asked whether their health behaviour had changed due to receiving their test results. In total, 27.3% (*n*=50) stated that it had and 72.7% (*n*=133) stated that it had not. Out of the 50 consumers who stated that their behaviour had changed, 45 (90%) described how it had changed.

All of the changes mentioned were either positive or neutral, and no cessation of any health behaviour was reported.

The changes in health behaviour mentioned and the percentage of respondents who mentioned each change are presented in Table 10. No total is included as some respondents gave more than one answer.

Table 10 Changes in Consumers' health behaviours

Change in Health Behaviour	Percentage (N)
Healthier diet	53.3 (24)
More exercise	26.7 (12)
Taking vitamins or supplements	13.3 (6)
Preventative checks such as eye tests	8.9 (4)
Looking into high risk items	6.7 (3)
Stopped or reduced caffeine intake	6.7 (3)
Lost weight	6.7 (3)
Generally more health conscious	6.7 (3)
Generally reducing risk conditions	4.4 (2)
Other	15.6 (7)

When asked whether their health anxiety had changed due to receiving their results, 24.6% (*n*=45) stated that it had and 75.4% (*n*=138) stated that it had not. Of the 45 respondents who stated that their health anxiety had changed, 75.6% (*n*=34) described how it had changed; 85.3% (*n*=29) stated that their anxiety had decreased, three individuals reported that it had increased and two stated that it had both increased and decreased. Most respondents who described how their health anxiety had changed reported that it had only changed by a small amount.

5.3.2 Differences in Health Behaviours and Anxiety Levels

5.3.2.1 Health-Behaviour Scores

The health-behaviour scores for the consumers and potential consumers groups are shown in Table 11 and Figure 12. Both a table and a figure are included to fully illustrate the distribution of the scores since a distribution-based significance test was used (as described below). As described in the Research Methodology (see section 4.3.3.4), the health-behaviour scores are from 0 to 6, representing the number of recommended health behaviours that each respondent reported following. The consumers' scores were generally higher than the potential consumers', with a mode of five and three and a mean of 4.18 and 3.64 respectively.

Table 11 Health-behaviour scores of the Consumers and Potential Consumers groups

Health-behaviour score	Consumers Percentage (N)	Potentials Consumers Percentage (N)
0	1.3 (2)	1.6 (1)
1	2.0 (3)	4.7 (3)
2	11.2 (17)	9.4 (6)
3	13.2 (20)	34.4 (22)
4	24.3 (37)	20.3 (13)
5	30.9 (47)	21.9 (14)
6	17.1 (26)	7.8 (5)
Total	100 (152)	100 (64)

A two-sample Kolmogorov-Smirnov test (which is based on cumulative frequencies) showed that there was a significant difference between the distribution of the health-behaviour scores between the two groups, with p=0.022.

The cumulative frequencies of the health-behaviour scores for the two groups are compared in Figure 13. This clearly shows that for each score on the chart (apart from 6) there is a higher cumulative frequency for the potential consumers, which means that a higher proportion of their scores are lower than the consumers group than vice versa.

Figure 12 The health-behaviour scores of the Consumers and Potential Consumers groups

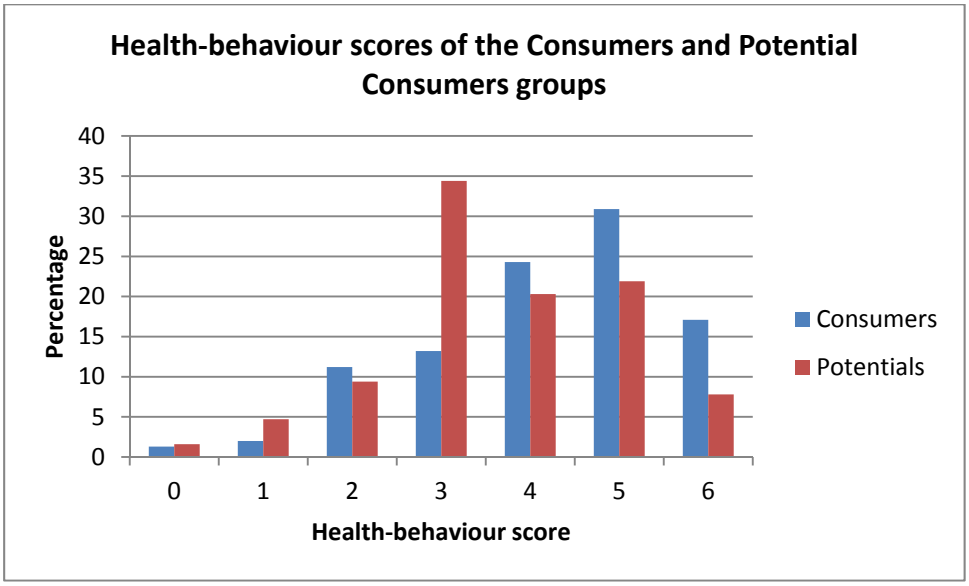
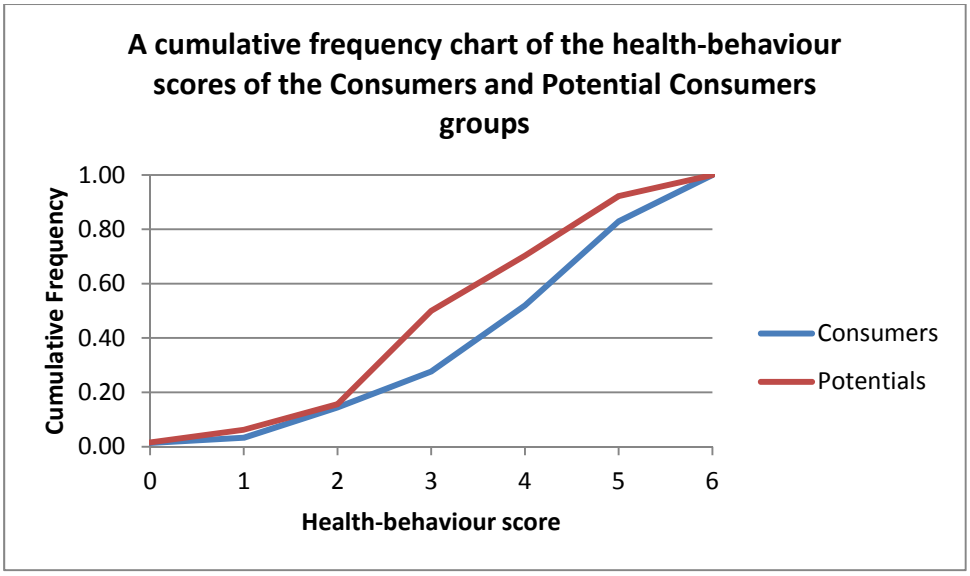


Figure 13 The cumulative frequencies of the health-behaviour scores of the Consumers and Potential Consumers groups



The scores were then split into two groups: a high-score group of respondents with scores of four or higher and a low-score group of respondents with scores of three or lower. A two-sided Pearson's Chi-Square test showed that there was a significant difference between the proportion of consumers and potential consumers in each score group $X^2(1, n = 216) = 10.01$, $p = 0.00$, with 72.4% of consumers and 50.0% of potential consumers in the high score group, compared with 27.6% of consumers and 50.0% of potential consumers in the low score group.

As described in section 4.3.3.1, 50 respondents stated that their only reasons for pursuing DTC genetic tests were genealogical. These respondents have been included in the analyses as, although they did not purchase the test for health reasons, they were still exposed to the health-risk information. However, in order to check whether their inclusion influenced the results, the analyses were repeated with these 50 respondents excluded.

The main results were: when comparing the total health-behaviour scores, the mean for the consumers was 4.25 and the mean for the potential consumers was 3.64. The mode for the consumers was five and the mode for the potential consumers was three. A two-sample Kolmogorov-Smirnov test found a significant difference between the distributions with p=0.030. When the scores were split into high and low scores, 73.3% of consumers had a high score compared with 50% of potential consumers. A Pearson's Chi-squared test found a significant difference between the two $x^2(1, N = 174) = 9.272$, $p = 0.002$.

Since the mean and mode health-behaviour scores were similar to the original scores with the genealogical respondents included, and since there was no difference in which results were and were not significant, it is reasonable to assume that the inclusion of respondents who stated that their only reason for purchasing the test was genealogy did not adversely affect the results.

5.3.2.2 Individual Health Behaviours

The percentage of respondents who were classified as normally adhering to each individual health behaviour (as described in section 4.3.3.4) is shown in Table 12. For both consumers and potential consumers the health behaviour normally adhered to by the highest percentage of respondents was smoking abstinence, with over 90% of respondents so doing. This was followed by sufficient fibre intake and sufficient exercise.

Table 12 Chi-squared tests for differences in individual health behaviours

Health behaviour	Consumers Percentage followed	Potential Consumers Percentage followed	Significance
Salt intake	63.7	55.1	Not significant $x^2(1, N = 240) = 1.557, p = 0.212$
Fat intake	57.1	50.7	Not significant $x^2(1, N = 250) = 0.850, p = 0.357$
Fibre intake	74.0	64.5	Not significant $x^2(1, N = 245) = 2.294, p = 0.130$
Fruit and vegetables	53.6	38.8	Significant $x^2(1, N = 261) = 4.890, p = 0.027$
Smoking	92.7	93.8	Not significant $x^2(1, N = 260) = 0.103, p = 0.749$
Exercise	66.3	61.0	Not significant $x^2(1, N = 249) = 0.639, p = 0.424$

With the exception of smoking abstinence (where the proportions were almost identical), for each health behaviour a higher percentage of the consumers group normally adhered to the health recommendations than the potential consumers group, although the only significant difference was for fruit and vegetable intake. A two-sided Pearson's Chi-Square test found a significant difference between the two groups in the percentage who normally had a sufficient fruit and vegetable intake $x^2(1, n = 261) = 4.89$, $p = 0.03$, but not in the percentage who normally had a moderate salt intake, a moderate fat intake, sufficient fibre intake, sufficient exercise or who were normally abstinent from smoking.

As in the previous section, the same analyses were also conducted with those who bought the test solely for genealogical reasons excluded. As above, this resulted in no differences for which results were significant and which not, adding further weight to the assumption that the results were not adversely affected by the inclusion of these respondents.

5.3.2.3 Health Anxiety

Respondents were asked two questions about health anxiety; one on general anxiety about health and the other on anxiety about developing a serious disease. For both questions respondents ranked their anxiety on a 10 point scale.

For general anxiety about health, the mean rating for the consumers was 3.80 and for the potential consumers was 4.22. The mode for both groups was two. A two-sample Kolmogorov-Smirnov test found no significant difference between the distributions of the two groups, with p=0.629.

For anxiety about developing a serious disease, the mean rating for the consumers was 3.52 and for the potential consumers was 3.92. The mode for both groups was two. A two-sample Kolmogorov-Smirnov test found no significant difference between the distributions of the two groups, with p=0.301.

5.3.2.4 Potential Consumers Group

As described in Methods, for the analyses of differences in health behaviour and anxiety, consumers who had not yet received their results were added to the potential consumers group. The potential consumers group therefore consisted of (out of those who had fully answered the questions) 45 'true' potential consumers and 19 consumers who had not yet received their results. These two groups were compared to identify any significant difference that might have affected the analysis.

The mean health-behaviour scores for the 'true' potential consumers was 3.67 and the mean of those who had not yet received their results was 3.58, a difference of 0.09. The mode for both groups was three, with almost identical standard deviation (1.37, 1.35). An independent samples t-test found no significant difference between the two (p=0.81).

5.3.2.5 Perceived Health Risk

Consumers were asked to give their impression, from their results, of their average risk of developing a serious disease. They were asked to rate this risk as one of five answers: a significantly lower than average risk of disease, a slightly lower than average risk of disease, an average risk of

disease, a slightly higher than average risk of disease and a significantly higher than average risk of disease.

Consumers' reported perception of their overall risk of serious disease based on their DTC genetic test results is shown in Table 13. The results follow a rough bell curve, with over half of consumers reporting an impression of an average risk of disease.

Table 13 Consumers' reported perceived risk of serious disease based on DTC genetic test results

Perceived Health Risk	Percentage (*n*)
Significantly lower than average risk of disease	2.9 (5)
Slightly lower than average risk of disease	26.3 (45)
Average risk of disease	52.0 (89)
Slightly higher than average risk of disease	14.0 (24)
Significantly higher than average risk of disease	4.7 (8)

The five different answer options were assigned a number from one to five on an ordinal scale. Using Spearman's Rank correlation, no significant correlation was found between perceived risk and health-behaviour score ($r = -0.08$, $n = 141$, $p = 0.36$, two-tailed), general anxiety about health ($r = 0.10$, $n = 165$, $p = 0.20$, two-tailed) or anxiety about developing a serious disease ($r = 0.15$, $n = 165$, $p = 0.06$, two-tailed).

Consumers were then split into two groups to compare the perceived health risks of those who stated that their behaviour had changed and those who stated that it had not. A two-sample Kolmogorov-Smirnov test found no significant difference between the two groups, with p=0.778.

5.3.2.6 Underlying Factors

Respondent demographic variables (age, proportion in the managerial and professional occupations socioeconomic category, gender, proportion of Caucasian ethnicity, education level, proportion residing in the USA) were analysed to determine whether they accounted for the difference in health-behaviour scores. A two-sided Fisher's Exact test found one significant difference between the consumers and potential consumers groups for age (p=0.00), with consumers significantly more likely to be in an older age group than potential consumers, but no significant difference for other demographic variables. However, no significant correlation was found between age group and behaviour scores (Spearman's r) : ($r = 0.130$, $N = 214$, $P = 0.057$, two-tailed).

The results were then weighted to take age group into account, and the health-behaviour scores compared again. A significant difference was still found between the health-behaviour scores of

consumers and potential consumers (p=0.03). The results were then weighted for the other demographic variables individually and the health-behaviour scores compared. A significant difference was still found between the health-behaviour scores of consumers and potential consumers when weighted for two socioeconomic categories (the managerial and professional occupations category versus the other categories combined) (p=0.04), ethnicity (Caucasian versus other ethnicities combined) (p=0.05), education level (p=0.03), gender (p=0.02) and country of residence (USA versus other countries combined) (p=0.01).

A stepwise multiple regression was then performed in an attempt to build a model of all predictor variables for the health-behaviour scores (n=137) i.e. all of the variables (such as age or gender) "that may be useful in predicting the scores" (Brace et al 2006, p.228). The results of this analysis are shown in Table 14. The only significant predictor variable was membership in consumers or potential consumers group (B = -0.48, SE B = -0.16, p = 0.03).

Table 14 Results of multiple regression analysis

Variable	Coefficient	*P*
Membership of consumers or potential consumers group	B = -0.48 SE B = -0.16	0.03
Gender	Beta In = -0.14	0.06
Education Level	Beta In = 0.12	0.09
Membership of managerial and professional occupations socioeconomic category	Beta In = -0.08	0.28
Residence in USA	Beta In = -0.06	0.39
Caucasian Ethnicity	Beta In = 0.05	0.47
Age Group	Beta In = 0.03	0.67
N	137	

5.3.2.7 Mean Website Assessment Scores

As described in the Research Methodology (see section 4.3.3.5), respondents were asked to rate their opinion of the website from which they purchased a DTC genetic test by indicating to what extent they agreed with six statements on a seven-point Likert scale. These results are shown in section 5.4 below. In order to assess whether or not respondents' opinions of the websites were correlated with their health behaviour or anxiety, a mean was taken of each respondent's answers to create a mean website-assessment score for each individual, which was then compared against their health-behaviour scores, anxiety about health in general and anxiety about developing a serious disease.

No significant correlation was found between mean website-assessment score and health-behaviour score (Spearman's r) : (r = 0.142, P = 0.084, two-tailed) or between mean website-assessment score and anxiety about developing a serious disease (Spearman's r) : (r = -0.147, , P = 0.051, two-tailed). There was a significant correlation between mean website-assessment score and anxiety about health in general (Spearman's r) : (r = --0.155, P = 0.040, two-tailed).

The mean website-assessment scores of consumers who stated that their behaviour had changed were compared with the scores of those who stated that their behaviour had not changed. For those whose behaviour had changed, the mean was 6.48 and for those whose behaviour had not changed, the mean was 6.25. A two-sample Kolmogorov-Smirnov test found no difference between the two groups, with p=0.451.

5.3.2.8 Discussion

Although causation can never be established from a cross-sectional survey such as this, the results point towards a positive effect of DTC genetic tests on the health behaviour of consumers; over a fifth reported a change in health behaviour and the health-behaviour scores of consumers were significantly better than those of potential consumers. Although there was only one significant difference between consumers and potential consumers with regard to individual health behaviours, the higher (but non-significant) adherence to each health behaviour except smoking abstinence likely resulted in the significant difference between overall health-behaviour scores. The extensive analysis of the demographic variables indicates that these results were not due to underlying demographic factors, and the repeat analysis of the results with respondents who had purchased a test solely for genealogical reasons excluded indicated that their inclusion did not influence the results. It is also important to note that no respondent mentioned any negative effects on health behaviour, such as the cessation of a healthy activity, which has been a prevalent concern in the literature.

The results for health anxiety are less clear than those for health behaviour. Many respondents did report a decrease in health anxiety, and some, albeit a small minority, reported an increase. However, there was no significant difference between the health anxiety scores of consumers and potential consumers. It is likely that this discrepancy is due to the nature of the changes to health anxiety; most reported changes were small, and so the comparison of consumers and potential consumers may not have been sensitive enough to detect them.

Although perceived health risk was not correlated with health behaviour or health-anxiety scores, it is interesting to note that there was a significant correlation between mean website-assessment scores and anxiety about health in general. This raises several interesting possibilities. For example, respondents' opinions about the websites may have been affected by their levels of health anxiety. Alternatively, the websites themselves may have influenced respondents' health anxiety, with those whose anxiety increased possessing a more negative view. Although an explanation cannot be ascertained with current data, it is a topic that would benefit from further research.

5.4 Website Assessment

5.4.1 Results

As described above and in the Research Methodology (see section 4.3.3.5), respondents were asked to assess the website from which they purchased a test by rating their agreement to six statements on a seven-point Likert scale. The scores given by consumers and potential consumers were compared to identify any significant differences between the two groups. A two-sample Kolmogorov-Smirnov test was used to compare the scores for each statement. The results are shown in Table 15.

Table 15 Differences in website assessment scores between the Consumers and Potential Consumers groups

Statement	Consumers Mean score	Potential Consumers Mean score	Significance
I am generally satisfied with the information provided on the website	5.75	5.30	Significant P=0.004
I had trouble understanding some of the information on the website	2.63	2.64	Not significant P=1.000
There is adequate information on the website to make a decision about buying a test	5.49	4.95	Significant p=0.013
The information on the website appears to be trustworthy	6.09	5.64	Significant P=0.050
The information on the website appears to be reliable	6.01	5.58	Significant P=0.031
I had to look at other sources to find enough information to make a decision about buying a test	3.37	3.82	Not significant P=0.488

For each statement the consumers' mean rating was better than that of the potential consumers i.e. higher for positive statements and lower for negative statements. There was a significant difference in the distribution of the scores for four of the six statements, with the most significant difference

for the statement 'I am generally satisfied with the information provided on the website', followed by 'there is adequate information on the website to make a decision about buying a test'.

5.4.2 Discussion

It is interesting to note that, as a whole, respondents gave a relatively good assessment of the website from which they purchased a test, considering them to be reasonably trustworthy with satisfactory levels of information. As this is the first piece of research to examine such matters from the consumers' point of view, it is interesting to compare their generally positive assessment with the generally negative assessment of the literature.

One notable finding was the significant difference between the website-assessment scores of consumers and potential consumers for four out of the six statements. This indicates that, as a whole, consumers and potential consumers felt differently about the information provided, with consumers generally possessing a more positive opinion. It is not possible to determine the reason for this difference, but it does raise the possibility that respondents' opinions of the websites affected their decision on whether or not to purchase a test.

5.5 Information Sources

5.5.1 Results

Respondents were asked where they had looked for information about DTC genetic tests. In total, 172 consumers answered the question correctly. Out of those, 153 (89%) stated where they had looked for information, 13 (7.6%) stated or strongly implied that they had not looked for information and 6 (3.5%) stated that they had not looked further for information or only listed in which tests they were interested. The sources of information used by consumers and the percentage of the 172 respondents who used each source are shown in Table 16. No total is included as some respondents gave more than one answer. The mode source of information was blogs, with over a fifth of respondents stating that they had used one. This was followed by Google and general internet use, with 18% each. Sources mentioned by only one respondent were included in the 'Other' category, and included documentaries, online support groups and YouTube.

The number of sources used by consumers that were not DTC genetic testing companies or their websites is shown in Table 17. The highest proportion of respondents (40.1%) only used one other source.

Table 16 Information sources used by Consumers

Source		Percentage (N)
Blogs:	total	22.7 (39)
	specifically scientific	9.3 (16)
	non-specific	13.4 (23)
Google		18.0 (31)
General internet use		18.0 (31)
Articles		11.0 (19)
Literature		10.5 (18)
DTC website: total		10.5 (18)
	comprehensive look	1.2 (2)
	unspecified	9.3 (16)
Forums:	total	7.6 (13)
	specifically scientific	5.2 (9)
	non-specific	2.3 (4)
General friends		7.0 (12)
Friends who are experts		6.4 (11)
Reviews		6.4 (11)
Mailing lists: total		5.8 (10)
	specifically scientific	3.5 (6)
	non-specific	2.3 (4)
Friends who have taken the test		5.8 (10)
DTC company		5.2 (9)
Wikipedia		5.2 (9)
Other named website		4.7 (8)
General reports		4.7 (8)
Twitter		4.1 (7)
Previous testing		2.9 (5)
Books		2.9 (5)
Experts not in the person		2.3 (4)
Workshop		1.7 (3)
Work		1.7 (3)
Magazines		1.7 (3)
ISOGG		1.2 (2)
General reading		1.2 (2)
Other		3.5 (6)

Table 17 Number of other sources used by Consumers

Number of other sources	Percentage (N)
0	13.4 (23)
1	40.1 (69)
2	27.3 (47)
3	15.1 (26)
4	3.5 (6)
5	0 (0)
6	0.6 (1)

A number of the sources were considered suitable sources (a source that is likely to give useful and reliable information about DTC genetic tests) for example: blogs, the literature, forums, mailing lists, documentaries, experts, workshops, the International Society of Genetic Genealogy (ISOGG) and online support groups. The analysis erred on the generous side i.e. all blogs, forums and mailing lists were considered suitable sources rather than just those which were specifically described as scientific- or genetics-based. The number of suitable sources used by consumers was then assessed. The results of this analysis are shown in Table 18.

Table 18 The number of suitable sources used by Consumers

Number of Suitable Sources	Percentage (N)
0	52.3 (90)
1	37.2 (64)
2	9.3 (16)
3	1.2 (2)

Based on their survey answers, over half of the respondents did not look at any suitable sources, whilst a further 37.2 percent only looked at one.

Respondents were asked what information they searched for when using the sources which they reported in the survey. In total, 45 consumers correctly included this information in their answer. The information that consumers searched for and the percentage that looked for each item are shown in Table 19. No total is included as some respondents gave more than one answer. The item searched for by the highest proportion of respondents (26.7%) was information about the coverage of the tests. This was followed closely by information about the cost of the tests, which was searched for by just under a quarter of respondents. Areas of information mentioned by only one respondent were included in the 'Other' category and included sample reports, bibliography, who is

associated with the company, information available elsewhere, existing data, interpretation, what has been learned so far, genetics of a specific disease, DTC genetic tests in general, ancestry information, publishing records of company scientists, method, support and company history. See Appendix I for explanations of category names.

When asked if they were satisfied with the information that they had found, 92.0% (183) of consumers were satisfied and 8.0% (16) were not satisfied. Just over half (55.3%) stated that this was the way that they would normally search for health information. When asked if they would recommend this way of searching for health information to other people, over two thirds (70.9%) stated that they would.

Table 19 Information Consumers searched for

Information searched for	Percentage (N)
Coverage	26.7 (12)
Cost	24.4 (11)
Tests	15.6 (7)
Reliability	11.1 (5)
Reviews	8.9 (4)
Information produced by tests	8.9 (4)
Usefulness	6.7 (3)
Technical details of analysis	6.7 (3)
Raw data or third party analysis	4.4 (2)
Other	31.1 (14)

5.5.2 Discussion

As stated previously, this research is the first to examine the informational aspects of DTC genetic tests from consumers' point of view. Therefore, the finding that most (86.6%) consumers used information sources other than the websites of the companies that sell the tests is vitally important when examined alongside the concerns in the literature about information provision; whatever the state of the information provided on the company websites the majority of consumers did not solely rely upon them.

However, although a wide variety of different sources were used, many of them could not be considered suitable sources with which to learn about healthcare information. When this was taken into account, it was estimated that just over half (52.3%) of consumers did not use a suitable source of information. This finding is worrying, and appears to support the aforementioned concerns, albeit with the caveat that it is an estimate based on survey answers rather than an in-depth analysis.

Another notable finding is that more than nine in 10 (92%) of consumers were satisfied with the information that they had found, and over two thirds (70.9%) would recommend their way of searching for information to other people. This illustrates that, whether or not it is considered that consumers have access to the information that they need, consumers themselves were generally happy with the information that they had found.

As stated above, a wide variety of sources were used by consumers. Once again blogs were the most commonly-used source. As in section 5.2.2, this may be due to the focus on social media in the sampling method, that the tests can only be purchased online or because of the large quantity of information about DTC genetic tests that can be found on blogs. It is interesting to note that the websites of the companies that sell the tests were only the joint fifth most commonly-mentioned source. However, some consumers may have assumed that the question referred to sources other than the company website, and so this finding should be taken with caution. Also notable is the fact that no respondent mentioned using the Genetics Home Reference website, which (as described in the section 2.2.3) was created for exactly this type of situation.

5.6 Information Need

5.6.1 Results

Respondents were asked what information they wished to know before purchasing a test. There were 161 usable responses from consumers and 51 from potential consumers. Twenty-one (13.0%) consumers and three (5.9%) potential consumers stated that there was no information that they wished to know beforehand. The information that respondents wished to know is shown in Table 20. No total is included in the table as some respondents gave more than one answer. The type of information that the highest proportion of consumers wished to know was information about the coverage of the tests, with just under a third of consumers stating this. In contrast, just under half (45.1%) of potential consumers stated that they wished to know information about accuracy, but less than a quarter (23.5) mentioned coverage. Types of information mentioned by only one respondent are included in the 'Other' category, this includes: transparency, downfalls, repeatability, company's years in business, third party tools, stability of company, ancestry examples, safety, type of testing, support and treatment they have. See Appendix I for explanations of category names.

Consumers were asked if there was any further information that they wished to know after receiving their results. Only 22 responses were usable, possibly due to confusion over the question. The results are shown in Table 21. Just under a third of consumers (31.8%) wished to know information about the interpretation of or further research into the raw data, and just under a

Table 20 Information that respondents in the Consumers and Potential Consumers groups wanted to know

Type of information	Consumers Percentage (N)	Potential Consumers Percentage (N)
Coverage	32.3 (52)	23.5 (12)
Accuracy	21.1 (34)	45.1 (23)
Cost	19.3 (31)	9.8 (5)
Privacy	16.8 (27)	13.7 (7)
Results	13.7 (22)	19.6 (10)
Sample	10.6 (17)	7.8 (4)
Analysis	9.3 (15)	9.8 (5)
Data	8.7 (14)	3.9 (2)
Trustworthy	7.5 (12)	0 (0)
Interface	6.8 (11)	11.8 (6)
Security	6.2 (10)	13.7 (7)
Confidentiality	5.0 (8)	11.8 (6)
Time	4.3 (7)	9.8 (5)
Interpretation	3.7 (6)	7.8 (4)
Sharing	3.1 (5)	2.0 (1)
Updates	3.1 (5)	2.0 (1)
Useful	2.5 (4)	3.9 (2)
Lab information	1.9 (3)	5.9 (3)
General	1.9 (3)	0 (0)
Company	1.9 (3)	3.9 (2)
Business model	1.9 (3)	0 (0)
Sample size	1.2 (2)	2.0 (1)
Comparisons	1.2 (2)	0 (0)
Site founders	1.2 (2)	0 (0)
Website	1.2 (2)	0 (0)
Prevention	0.6 (1)	2.0 (1)
Users	0.6 (1)	2.0 (1)
Ownership	0 (0)	3.9 (2)
Other	5.0 (8)	9.8 (5)

Table 21 Information Consumers wanted to know after receiving their results

Categories	Percentage
Interpretation/research into raw data	31.8 (7)
Analysis	22.7 (5)
Accuracy	18.2 (4)
Prevention Strategies	9.1 (2)
Other	31.8 (7)

quarter (22.7%) information about the analysis. Types of information mentioned by only one respondent were included in the 'Other' category, these included: doctor, coverage, meaning of words, application, clinically actionable, gene/environment interaction and reuse sample. See Appendix I for explanations of category names.

5.6.2 Discussion

Respondents identified a wide range of information that they wished to know before purchasing a test. It is not surprising that coverage and accuracy were the two most sought-after areas of information; it is obviously important for consumers to know what the test covers and how accurate it is. What is interesting is the difference between consumers and potential consumers for these two areas of information. The most commonly-mentioned area for consumers was coverage, which was mentioned by 50% more respondents than accuracy. In contrast, accuracy was the most important area for potential consumers; mentioned by almost double the number of respondents who mentioned coverage. Although there is not enough data to explain this difference, it is possible that it is due to an underlying difference of opinions about the tests; since accuracy was more important to potential consumers it is possible that the reason they have not yet purchased a test is due to concerns in this area.

As in other parts of the informational aspects of the tests (and as frequently mentioned), this research is the first to examine information needs from the consumers' point of view. Therefore the large list of different needs generated is useful, and provides a good comparison with the types of information that professional organisations and researchers think consumers need to know (see Research Methodology and Content Analysis).

5.7 Summary

This survey has broken considerable new ground in several ways, being, to the author's knowledge, the first study to include 'real' consumers of DTC genetic tests (i.e. individuals who have sought out and purchased a test themselves) whilst remaining independent of providers of DTC genetic tests, to include potential consumers i.e. individuals who are similar to consumers but have not yet received any test results and to examine the informational aspects of DTC genetic tests from the consumers' point of view.

It has also provided a large amount of useful data. Although the cross-sectional nature of the survey precludes the establishment of causation, the results point towards a positive impact of DTC genetic tests on the health behaviour of a minority of consumers. Importantly, no adverse effects of the

tests on health behaviour were mentioned by any consumer. The findings in relation to health anxiety were less clear, with reported effects (mainly positive but some negative) but no significant difference between consumers and potential consumers. The results also began to uncover aspects of respondents' information behaviour. For example, a large number of information needs were identified, the most common of which were to do with the coverage or accuracy of the tests. Consumers reported using a large variety of different sources of information, with blogs identified as the most commonly-used source. However, just over half of consumers did not report using any suitable sources. Finally, respondents gave a relatively good assessment of the information provided on the website of the company from which they purchased, or were thinking of purchasing, a test, considering them to generally give satisfactory levels of information and be reasonably trustworthy.

6 Email Interviews

This chapter presents the results and analysis of the email interviews[4]. It is organised into nine different topics which were discussed in the interviews: behaviour, anxiety, health professionals, surprise, reason for purchase, information, understanding, genetic knowledge and other or anything to add. In total, responses were gathered from 36 respondents. However, some respondents were not asked every question (see section 4.4.2.1) and, due to the nature of email interviews, not every respondent replied to every question. Therefore the respondents described in each section refer only to those respondents who answered the relevant question or questions, not to the respondents in full.

6.1 Behaviour

6.1.1 Change in Behaviour

In total, 17 out of 33 respondents made changes to their health behaviour as a result of DTC genetic test results.

6.1.1.1 Type of Change

Respondents reported a variety of different changes to their health behaviour after receiving their results. The most commonly-reported change was to diet, with 14 respondents so reporting. However, this category covers a wide variety of different dietary changes. These include some that were small, specific changes, but most involved more than one change, with some respondents reporting comprehensive changes to their diet. For example:

> *My genetic scan showed a high risk for colorectal cancer. Solely because of this, I am now eating more fiber.* [20]

> *I have oatmeal with other grains 6 mornings a week and eat a vegetarian almost vegan diet.* [7]

Out of the 14 respondents who changed their diet, only one changed diet alone. Seven respondents reported making changes to two areas, diet and exercise. Two respondents reported making changes to diet and weight. Four respondents reported changes to several areas.

[4] All included quotes are copied verbatim from the email interview transcripts.

I DO try to improve overall 'life=style' patterns, like watching my sugar intake, try to exercise more, etc. [12]

I have reduced my intake of caffeine and also lost a stone in weight. [22]

After seeing my highest genetic risks were heart related and psoriasis I have been going to a skin doctor ever 6 months for a check ... I also started doing more exercise and eating a healthier diet. [7]

Exercise itself was the second most reported change of behaviour, with 11 respondents so reporting. However, no respondent stated that they had only changed their exercise; all of the respondents who had changed their exercise had also changed their diet, and some had changed other areas as well.

...[risks can be handled by] only small amounts of sugar/salt/fat, more fruit/greens etc. a bit more pulse related exercise doing the day (ex. spinning, 1hour a day). [13]

Changes to exercise included large changes such as the use of personal trainers and smaller changes such as modifying an existing routine.

I was already doing weight lifting for exercise, but I added cardiovascular exercise to my routine based on various studies looking at the effects of that type of exercise of Type 2 diabetes, cardiovascular diseases, etc. [3]

Seven respondents mentioned weight; six that they had changed their behaviour in relation to it (i.e. decreased weight) and one who was making an effort to keep his weight within the normal BMI range. Perhaps unsurprisingly, all of these respondents also mentioned changes to diet, and three mentioned changes to exercise.

I am overweight and have high cholesterol ... I need to get the weight off my back and lower my cholesterol and triglycerides ... I need to increase my exercise. I have gone back and forth but I have not given up. I have lost 20 lbs but need to lose much more. I have been substituting Stevia for sugar in a lot of things. Have upped my veggie and fruit intake. I need to work more on portion sizes. [35]

Six respondents reported a change involving medical tests. For four of these respondents, this change was reported alongside other changes to health behaviour. The medical tests for three of these four were measuring blood pressure at home, six-monthly check-ups with a doctor for

psoriasis and a five-yearly check for hemochromatosis. The fourth respondent reported that he needed to increase his consumption of B vitamins in order to keep a normal homocysteine level. He changed his diet and stated:

> *I also did lab tests which confirmed that I had high homocysteine which reduced a few weeks after diet changes, as did levels of DNA damage.* [31]

Out of the other two remaining respondents who reported medical tests, one also was tested for hemochromatosis. The other respondent described how she had been diagnosed with breast cancer after pushing for a biopsy because of her test results.

> *That's just it, i started paying more attention to when i started feeling a bit off, my appetite decreased, and i started feeling a bit more tired, this wasn't normal for me, i also lost weight just a few pounds but my loss of appetite was also accompanied by some gastro discomfort, diarrhea, so when i felt off i took note of it and brought it up to my general practioner during my annual exam, she gave me the referral to the mammogram, a well as ordered blood work, which indicated i was low on vitamin D, when i went for the mammogram i was still feeling off a bit, i was called a week later asked to return to repeat a mammogram and then was told by the radiologist afterwards that i had 2 micro calcifications and to return in 6 months for a repeat to see if they increase in size and need to be removed, i said No way, my Dna indicates i am a high risk of Breast cancer and i recently found close relatives with other forms of cancer,i want a biopsy now even if i have to put it on my american express...a week later i was biopsied.* [10]

Other reported changes to health behaviour included an increased amount of sleep reported by one respondent, along with the playing of word games, an increased consumption of tea and consumption of omega 3 supplements for the purpose of prevention of Alzheimer's disease. Another respondent reported a recognition that their sleep should be better. A third described a change in medication alongside other behavioural changes mentioned above. Two respondents were identified as carriers of cystic fibrosis, and, amongst other behavioural changes (non-related to cystic fibrosis), have either encouraged family members (and others) to test or will do when they want to have children. One stated:

> *I also found that I am a carrier of cystic fibrosis. Because of this, I have encouraged my partner and family members to test. I am also much more aware of any studies in this regard and read any information that comes out about CF. My awareness of Mendelian*

Diseases is heightened and I encourage women of child bearing age to test as a result. [20]

Some of the respondents mentioned that some behavioural changes were difficult to make.

My difficult point is sugar, where I still allow myself a bit in the evening, but soft drinks I've dropped. Also as I have pains in my back and joints due to an old damage, then I seldom get as much sleep as I should, so stress and sleep could be better factors as well. [13]

Many respondents also mentioned the success that they had had, or reported that the changes had lasted. For example one stated "*I feel that I just keep getting better about both my diet and exercise*". [7]

6.1.1.2 Reason for Change

The interviews highlighted several different reasons for respondents' behavioural changes.

The most common reason, reported by 12 of the 17 respondents, was that specific high risk areas of their results had prompted them to make specific changes to their behaviour. For example:

The test suggested that I was at increased risk of developing Type 2 Diabetes, Atrial Fibrillation and a couple of other things. I also had an increased sensitivity to Warfarin and I was a slow metabolizer of caffeine. Since then I have reduced my intake of caffeine and also lost a stone in weight. ***[Question: so were all of your changes in health behaviour linked to specific high risk areas in your results?]*** *Yes I suppose they were linked to specific high risk areas. There were some others that I couldn't do anything about.* [22]

One respondent described how she thought that specific areas of her results had caused her to change her behaviour.

I think the very 'act' of seeking a genetic report shows a level of prior interest in knowing more about one's health. However, once getting the report, I think that I was therefore a bit better informed, and with more information I became more aware of SPECIFIC areas of my health that I needed to be 'more alert' of my personal contribution to possible problems. [12]

For some respondents these specific risks matched what they knew of their family history. One respondent described how, for the three main diseases that she was at an increased risk for, either one of both of her parents had suffered from each of them. Despite knowing this family history, she would not have made any changes now without the test results. Another respondent described a similar situation in detail:

> *My genotyping results indicated I had an increased risk for developing Type 2 diabetes. I had already suspected it based on family history since both my mother and grandmother developed the disease. But, seeing the actual mutation and having access to the research showing how my genotype increased risk made it a bit more concerning to me. So, I have changed my diet and altered my exercise routine to help reduce my risk of diabetes.* [3]

One respondent mentioned that, prior to receiving his results, he was already aware that it was a good idea to make the changes to health behaviour that he made after receiving his results. However, he was spurred into action by several results that showed an increased risk, and by the fact that he had relations who had lived to an old age i.e.there was nothing in his results to stop him from doing the same if he counteracted the high risks.

Another respondent described how the decision to make changes was a mixture of specific results and that she was already thinking of making changes.

> ***[When asked how much of the changes were due to her results]*** *At a wild guess I'd say 50% but really, it's pretty hard to give an accurate assessment. With change, I find that it can be something small that pushes a change I'm thinking about making into action. In this case, the fact that I ordered the test was already part of thinking of changes.* [33]

One respondent described how her increased risk of heart disease had motivated to do more exercise.

> *I have always been active but I have been trying to do more exercise since I saw heart problems were my biggest risk. I do think about it more and try to push my self to get the heart rate up. I was getting a little lazy about exercising and this just reminded me I better do it.* [7]

Two respondents described how comparisons of the results with their family history made them decide to change their behaviour.

One of these respondents, who also made changes due to specific results (as described above), described how the comparison of her type two diabetes risk with her brother made her decide to exercise more, and also how she pays attention to risks running in the family.

> *My major risk is diabetes 2. My brother who was also tested shows an even HIGHER risk. I DO have diabetes 2. He does NOT. The difference in our lifestyles really is only one aspect: EXERCISE.... HE plays tennis twice a week, I am a 'computer-potato'..... The result? I am now attempting to exercise more.*
>
> *Several of my risks, like kidney disease, stroke, and heart disease I have 'running' in my family, and I pay closer attention to those areas as well, since the 'risk' is confirmed genetically.* [12]

The other respondent described how a comparison of his results with his mother's and grandmother's results (for whom he had also purchased a test) and his family history resulted in his decision to change his behaviour. He described how there was a large difference in life span of those in his family who had been careful with their health and those who had not, and he could see how some of his areas of high risks matched his family history. Even though these were not hugely increased risks, he could see from his family history how important they were.

> *....I have a few higher risk factors, and while it is not % by much, I can then see by what relatives have died of or their health as such, that these factors however small they might seem, can come forth if one is not carefull.*

Equally importantly, his results made him realise the parts of his family history for which he was at risk.

> *.... I think the testing was rather important in regards to the changes I took, as the health risks I found corresponded & fitted with the health & death records among close relatives.*

This allowed him to determine on which health issues he should be focusing.

> *... What changed after the test, was that I via comparing with family lore & own health far better could pinpoint where I relatively easy could set in preventively, in regards to a number of things that else was likely to set in with age, and since I'm only 40, I then still have the time to make a difference.*
>
> *In short, where I mostly had to make evaluated guesses before based on generel health advices, I can now narrow it down to a few specific items*. [13]

Two respondents described how their results led to a general realisation that they needed to improve their health.

One of these respondents described in detail how the investigation of three areas of his results (hemochromatosis, carrier of cystic fibrosis and increased risk of age-related macular degeneration), including appointments with his general practitioner, clinical geneticist and others, made him realise that he was in charge of his own health. This realisation caused him to change areas of his lifestyle that were unrelated to these results, in an effort to improve his general health; he therefore changed his diet, lost weight and increased his level of exercise.

> *I didn't make lifestyle changes because I had any particular risk factors. It was just that the results got me thinking about my health, and what I could do to reduce my risk of developing diseases.* [25]

The other respondent described how part of her reason for changing her health behaviour was due to specific results, and another part was due to a realisation after receiving her results of the importance of living healthily.

> *I guess the short answer to your question is that changes are for mixed reasons. I realize that as I age, my body changes and I'd better do something to keep as many quality-of-life items as I can. It was a real eyeopener to look at my genetics and really realize that what my doctors had been saying was true.* [6]

Two respondents mentioned that, as well as responding to specific areas of high risk, their decision to change their behaviour was also made out of a desire to generally protect their health. For example, when one was asked to clarify whether or not all of his behavioural changes were linked to specific high risk areas in his results, he replied "*yes, other than basic desire to improve health and live longer."* [26]

One respondent described how the behavioural changes she had made was due to a combination of her genetic test results and her doctor's advice on changes she should make for an existing medical condition. When asked the proportion of the changes that were due to each she stated "*50% due to genetic tests -- 50% due to doctor (surgery - test results...)"*, and when asked specifically about the tests:

> M*y Dad had 4 heart attacks and died from cancer, my grandfather (maternal) died of a heart attack very young so seeing the results of my chances at one made me re-think what I was doing to my body. So, it had a pretty heavy impact I would say.* [35]

Section 6.1.1.1 describes how one respondent decided to push for a biopsy when two microcalcifications were found in a mammogram. When asked if she had made any behavioural changes due to the test she replied "*no , but it did make me more in tune with my health"*. [10] The respondent described how it was this effect, along with the breast cancer risk highlighted in the results, which allowed her to be diagnosed much earlier than otherwise.

6.1.2 No Change in Behaviour

In total, 16 out of 33 respondents did not make any changes to their health behaviour because of their DTC genetic test results.

Six of the respondents described how they did not make any changes to their health behaviour because there was no cause for concern in their results.

> *There was nothing in my results that was of any particular concern. After spending some time trying to understand the statistics I realised that the predictions weren't particularly meaningful. Knowing that I supposedly have a marginally higher risk than the average for handful of obscure conditions doesn't really tell me very much.* [17]

Five respondents stated that there were no surprises in their results, either generally or because they knew their family history.

> *The simplest way to put it is that there was nothing in my genetic profile that surprised me. Doing a family history would give you the same basic information about me that my genetic test results did. It came as no surprise that I have higher predispositions for diabetes, high blood pressure, various cancers, heart attack, and Alzheimer disease (I have one copy of the APOE ε4 gene) etc. The only thing the genetic testing did was further confirm basically what I already knew*. [8]

Five respondents mentioned changes to their behaviour that were unrelated to their test results.

> *Yes, things have changed a lot in the past five months. I cannot make a causal connection to signing up for 23andMe, it's just what I do. I've bought a very nice road bike and take it to work every Friday, sometimes more often. That's a total of 30 miles of hills, about two hours of biking. I still hike weekly and SCUBA monthly as I did before 23andMe. I've also read a lot about caloric restriction and intermittent fasting diets, and have been eating every other day since mid-December. Every lunch meal, I either*

complete or begin a fasting period until the next lunch. On days I exercise, I do not fast. I chose this to improve my longevity and health now that I am an adult (25 years old). [11]

Four respondents mentioned that they already followed appropriate behaviour.

Previous to the test my level of exercise and diet were good and therefore I didn´t change that. The only thing that could raise some worry was an slighty elevated risk of age related macular degeneration. I consulted and ophtamologist who recommended antioxidant suplements that I was taking anyway. [18]

Four respondents described how their results had not shown any need for taking action.

I didn't change any health behaviour essentially because I didn't get any risks that I could associate with a change in living habits. [1]

Two respondents raised doubts over the usefulness of the results.

Also a lot of the data that 23andMe used related to the American population and it was not clear how much of this applied to the UK population and me in particular, and especially their predictions for diabetes and obesity. [17].

One respondent mentioned that she did not like a particular healthy behaviour, stating "*I get bored with exercise - find a way to make it interesting, and I'd do more*." [2]

One respondent described how, due to her knowledge of genetics, she did not see the need to change her behaviour.

Last may I finished my masters in genetic counseling. My knowledge of genetics and GWAS ***[(genome-wide association studies)]*** *I think helped quell any health concerns. I know that the strength and reproduciblity of GWAS studies are very low compared to traditional Mendelian genetic traits and conditions, so they don't hold so much weight in my opinion.* [36]

Lastly, one respondent described how he was unable to change his behaviour due to a medical condition.

Because I had/have AF (arterial fibrillation), both before and after the tests I was/am unable to change my exercise habits. [9]

6.2 Anxiety

6.2.1 Change to Anxiety

In total, 13 respondents stated that their health anxiety changed after receiving their results.

6.2.1.1 Decreased Anxiety

Eleven respondents stated that their anxiety had decreased after receiving their results.

Eight stated that their anxiety had stayed at the decreased level (although the other three did not state that it had increased again).

> *With the results I have in hand, I would say permanently reduced. Of course you never know when a new study will come along, LOL!! I still feel I'd rather know my risks than not. After all, ignorance does not negate risk. In other words: ignorance is not bliss.* [19]

Various reasons were given for respondents' decrease in health anxiety. The joint most common reason, cited by four respondents, was that their results had not shown an increased risk for diseases for which they had a family history. One respondent described how her grandmother and uncle had both suffered from Alzheimer's disease, and so she was happy that she did not have an increased risk for it. Members of her family had also been tested, and she was relieved that her husband had not inherited the BRAC mutations that increase risk of breast cancer from his mother (and so would not pass it on to their children) and that her son had not inherited a carrier status that both she and her daughter-in-law had. However, her anxiety was not increased by an increased risk for heart disease and diabetes in her results, as this matched with her family history and so she had been expecting it.

Two other respondents mentioned similar results; one found that he did not have a version of a tumour suppression gene that ran in the family and the other that he was at a decreased risk for prostate cancer, Alzheimer's disease and Parkinson's disease for which he had a family history (alongside a decreased risk for other chronic diseases). However, although this second respondent recognised that these results did not preclude him from contracting any serious disease, his anxiety was still decreased.

The fourth respondent mentioned that she was relieved that she did not have a higher risk for early-onset breast cancer and Alzheimer's disease, both conditions for which she had a family history, although she recognised that this did not mean that she would definitely not contract the diseases. She described how it was a combination of this information and a non-related medical diagnosis

which decreased her health anxiety, rather than just one alone.

> *Much of the change in anxiety levels was due to the negative results you mentioned [early-onset breast cancer and Alzheimer's]. But also, at about the same time, I had received a medical diagnosis which explained why I had suffered from nervous symptoms (which I thought of as hypochondria) for much of my life. The whole picture came together and the upshot was that my levels of anxiety changed. However this was not necessarily because of the DNA results alone. but a combination of circumstances.* [23]

The other joint most common reason for a decrease in health anxiety, cited by four respondents, was that they were now more knowledgeable about their health.

Two respondents described how it was simply knowing their genetics and what conditions they were at an increased risk for that had decreased their anxiety. One stated that he had "*more knowledge, more conditions to understand and ameliorate."* [11]

Another described how, since she had been adopted, she had not known any of her family medical history. Therefore, the results had allowed her to know which areas of her health to focus on, and had reassured her that she was not at a hugely increased risk of a particular disease.

The fourth respondent described how it was not a decreased risk that had decreased her anxiety, but that her results had matched what she knew about her family history, that she was not at a significantly increased risk for any fatal diseases and that she now knows what her risks are.

Three other reasons were given by respondents for a decrease in their health anxiety.

One mentioned that his anxiety had decreased a small amount because all of the risks in his results were for preventable diseases.

The second respondent described how he could combine his results and family history to work out which areas of his health to focus on. Alongside the information about interventions on the company website, this allowed him to decide what to do to prevent disease. This respondent also mentioned that since he had relatives who had lived to healthy lives to an old age he was hopefully about using this information to do the same himself.

The third respondent mentioned how his anxiety had actually increased after seeing his risk levels, but dropped down below the original level when he had decided on a course of preventative action.

i [sic] would say my anxiety certainly increased after learning my probability of developing genetic conditions, then decreased after i made some positive life changed. i would say my overall level of anxiety is lower than it was before i got the test, i sleep alot better at night knowing my chances and that im decreasing them. [26]

6.2.1.2 Both Increased and Decreased Anxiety

Two respondents described how their health anxiety had both increased and decreased due to their results.

One of these respondents stated that in the main his anxiety decreased, and his overall level of anxiety was lower now than before testing. This was because his risks were generally low, and that it was relieving to have actual information about his risks rather than speculation based on family history. However, he had been caused some anxiety by learning of his increased risk for some conditions. This had mainly been relieved by the knowledge that those conditions are preventable if the correct interventions are undertaken and treatable if contracted. However the anxiety had not been completely relieved.

The second respondent described how his anxiety had been changed by receiving his results.

Previous to taking the genetic test, I tried to avoid thinking about my health. Most of the time I ignored it, but then when I did think about it it would sent me into a panic, and I frequently become depressed. Since taking the test, and the resulting changes to my life, I think about my health far more (every time I choose food or plan activities). I suppose this generates a low buzz of mild (but manageable) anxiety, as opposed to the less frequency but less manageable large bursts of anxiety that I felt previously. In general I think it is better this way. [25]

6.2.2 No Change to Anxiety

In total, 17 respondents stated that their health anxiety did not change after receiving their results. A wide variety of reasons and explanations were given for this, with some respondents citing a long list of reasons for why their anxiety did not change.

The most commonly-given reason was respondents' general attitudes to health, attitudes towards the tests or knowledge about the tests. One respondent described how she was a level-headed, practical person who was action-oriented and so would do what she could to reduce her risk of disease. She also stated that she knew that the tests need to be taken 'with a grain of salt', that the

science is still uncertain and that 'everyone will show increased risk for something' [20]. Another respondent described how he works for an organisation involved in medical research and so he understands enough about the tests not to be caused anxiety. Similarly, a respondent who had recently completed a master's degree in genetic counselling stated:

> *My knowledge of genetics and GWAS I think helped quell any health concerns. I know that the strength and reproduciblity of GWAS studies are very low compared to traditional Mendelian genetic traits and conditions, so they don't hold so much weight in my opinion.* [36]

Two respondents stated that they just did not have health anxiety, and one stated that he knew genetics was just one variable in health. One respondent stated that everyone always faces risks, and as long as appropriate action is taken to reduce genetic risks, then they should not be dwelt upon. One respondent described how an understanding of statistics had stopped her being too worried about her results. The final respondent in this group stated that he indulged in activities that were unhelpful with regard to his increased risks, and if he ended up contracting a disease he would not blame his DNA.

One respondent stated that she had not had any anxiety since her parents lived to an old age and another respondent described how her high risk areas did not correlate with her family history. Conversely, four respondents stated that although they had areas of increased risk, they matched what they knew of their family history and so there were no surprises.

> *The simplest way to put it is that there was nothing in my genetic profile that surprised me. Doing a family history would give you the same basic information about me that my genetic test results did. It came as no surprise that I have higher predispositions for diabetes, high blood pressure, various cancers, heart attack, and Alzheimer disease (I have one copy of the APOE ε4 gene) etc. The only thing the genetic testing did was further confirm basically what I already knew.* [8]

Two respondents did not have any results which gave them a cause for concern i.e. high risks with a strong evidence base, and a third had already had the diseases for which he was at an increased risk. A fourth found that she was a carrier of cystic fibrosis, but was not worried as she was not planning on having any more children. Another respondent did have an area of high risk, but one for which there is no known preventative behaviour.

Finally, one respondent stated that she already lived a healthy lifestyle.

I am still not anxious about my health results since heart risk were my highest and I do everything I can to keep healthy. I'm active not over weight and a vegetarian (almost vegan) so I think that makes up for genetic risk. [7]

6.3 Health Professional

6.3.1 Shared Results

In total, 17 respondents had shared their results with at least one health professional. Many of these shared specific results with relevant health professionals, and one respondent reported sharing a particular result that was relevant to symptoms she was experiencing. Other respondents shared their results in general, or shared specific parts with one health professional (such as an increased risk of Macular Degeneration with their ophthalmologist) and their whole results with their general practitioner. One respondent mentioned sharing his results only because he'd been asked about his family history.

Respondents received varying reactions from health professionals, with some respondents receiving different reactions from each health professional with whom they had shared their results. The most common responses were positive, with 10 respondents reporting that they had shared their results with a health professional who was interested or who gave an otherwise good response.

My gastroenterologist was interested in the results and told me my colon cancer genetics were exactly what she would have expected given my colonoscopy results. [5]

[Ophthalmologist] *didnt know about the test. I explained it to her. good attitude and on my request gave me possible preventive measures for ARMD* ***[(age-related macular degeneration)]****.* [18]

Conversely, six respondents shared their results with health professionals who were indifferent or not interested. For example, one respondent stated "w*hen I took my health report from 23andme to my doctors, they were basically NOT interested".* [12]

Three respondents reported an unhelpful response from the health professional with whom they had shared their results.

...and a few of them ***[doctors]*** *directly denies that it can be used for anything, as they put it in same cat. as new-age crystals etc.* [13]

One respondent reported that although he had had a good response, one of the health professionals to whom he talked had not been keen on the idea of the general public having access to such information.

Only one respondent stated that his health professional was knowledgeable about genetics. The respondent's doctor knew his family history and knew enough about genetics to use this to isolate the important mutations from his results. Two respondents also reported that their health professionals were unsurprised at their results given their family history. However, four respondents had shared their results with a health professional who they thought was not knowledgeable enough about the topic. One stated "*I don't think my doctor really gave it much thought. I doubt she knew much about it*". [27]

Two respondents thought that their health professionals were intimidated by the tests. However, two reported that their health professionals were interested in buying a test themselves.

Four respondents were either referred for tests based on their results, or had their treatment changed. As described in section 6.1.1.1, one respondent was referred for a breast cancer biopsy which resulted in a diagnosis of breast cancer. Another respondent was referred to a genetic counsellor along with several tests to analyse high-risk areas of his results. The third respondent had a single ferratin test ordered based on her results, and the fourth respondent's medication was changed based on the drug response results.

One respondent was recommended (on request) preventative measures to follow. Two respondents reported that their health professional would keep an eye out for early-warning signs of the disease for which they were at an increased risk, although one said that they would have done so anyway:

> *He also said we would pay attention to potential markers of diabetes, but he would have done that anyway given my family history.* [3]

6.3.2 Not Shared

In total, 10 respondents stated that they had not shared any of their results with a healthcare professional. However, nine of them indicated that they were not against the idea.

Five respondents thought that they would share their results at some time in the future. One was specifically changing his general practitioner in order to do so.

> ***[When asked if he had shared his results]*** *I have not, mostly because my doctor seemed very disinterested! i am activly looking for a new doctor as a result. one who would take*

the time to look them over and advise me on other changes if needed. i delieve communicating these results to a medical professional is a very good idea. [26]

Three respondents reported that they had not shared their results because they did not seem to be currently important.

No I haven't shared any of my results with my doctor. I did wonder about sharing my drug response reports with my GP to see if he wanted to add them to my medical record. My results show that I potentially have a reduced response to hepatitis C treatment and an increased response to warfarin. However, I'm unlikely ever to need such treatment so it didn't seem worth the effort. [17]

One respondent stated that she thought that health professionals would be dismissive and disapprove of the tests, but that she would share her results if it became important to do so.

6.3.3 Opinions

Respondents were asked their opinions about individuals sharing their results with a healthcare professional. Their responses can be broadly grouped into two areas: opinions about sharing results and opinions about healthcare professionals in relation to DTC genetic test results.

6.3.3.1 Sharing

Seven respondents thought that consumers should share all of their results with a healthcare professional, and one respondent thought that it was a good idea to discuss results as doctors prefer to have full information about their patients. One respondent thought that consumers should be able to share results if they want to, and one stated that if in doubt consumers should always opt to share more. Five thought that consumers should share if there is an important reason to do so, but were either unsure about or against their otherwise sharing.

My response is that it is situational. For example, a specific issue like macular degeneration, where it was highest on the list and I would benefit from a more thorough look into the eye by the doctor, went well. On the other hand, primary care physicains are NOT equipped or educated to deal with a list of "possible" risk factors and the probablity. I wouldn't bore/bother them with this. If you had symptoms of a specific problem OR had a risk that could benefit from further tests such as the specific genes for Breast Cancer, then I would share that limited information. [2]

One respondent thought that it is normally appropriate to casually discuss results but no more. However, she made an exception for drug response results and carrier status if planning on having children, which she thought should always be shared. Three respondents stated that they were not against the idea of consumers sharing results, with two mentioning that this was on the condition of there being no resulting genetic discrimination.

Three respondents stated that it was up to the individual concerned; one mentioned that confidentiality is very important and that respondents can always ask a doctor if they are worried about a particular result.

One respondent raised doubts over doctors' ability to use the results, but stated that consumers could always share results and "*hope for the best*" [13].

Two respondents specifically stated that consumers shouldn't share all of their results with healthcare professionals.

6.3.3.2 Healthcare Professionals

The majority of opinions in relation to healthcare professionals and DTC genetic tests were negative. Ten respondents stated that healthcare professionals would have difficulty using the results, would have difficulty interpreting results, are out of date, are not equipped to deal with the results or similar sentiments. As one stated: "m*y opinion is that the medical profession don't really understand how these tests work".* [17]

Two respondents thought that it might be unnerving for some healthcare professionals to have knowledgeable patients, and pointed out that patients taking their own initiative are not always appreciated.

One respondent thought that some healthcare professionals might not be open to new things. Another stated that some may not take the tests seriously and a third mentioned that they may not be interested in little-known genetic associations.

One respondent warned about how healthcare professionals might react to the results.

> *Some do nothing with the recommendations and others swing to over treatment and even referrals to genetic counseling because of their discomfort with the results.* [36]

Two respondents stated that other sources may be more useful for patients.

My opinion is that the medical profession don't really understand how these tests work, and people are far more likely to get an intelligent and helpful response from one of our ISOGG (International Society of Genetic Genealogy) mailing lists. [17]

6.4 Surprise

Respondents were asked if they were surprised by their results. The highest number, 13, were either not surprised or not particularly surprised.

I was not too surprised by the results. I knew that by chance I would be at risk for some assortment of issues. [14]

Most of these respondents simply stated that they did not have any surprising results. However, three specifically stated that, although their results had shown an increased risk, because their risks matched their family history they were either unsurprised or not completely surprised.

I wasn't expecting anything in special, so I wasn't surprised per se. My most elevated risk compared to average (prostate cancer) is not completely surprising; my grandfather died of complications following a prostate cancer (getting a late diagnostic of prostate cancer at 80 is never good). But I didn't expect any special risk in that area. [1]

Eight respondents reported that they were surprised at their results. Six of these described how they were surprised at having an increased risk for an unexpected condition.

I was surprised to see that I have an increased risk for Bipolar Disease since I am an extremely even person who does not suffer from depression or manic episodes at all. [20]

Yes i was surprised at the Alzheimer thing as no-on in my family had anything like that and all my grandparents lived to late 70's and 80's - dad was 88 and mum's 90 (although she has memory problems and slight dementia it is not Alzheimers. [22]

Interestingly, all of these six people had changed their behaviour due to receiving their results.

Two respondents were surprised at being a carrier of cystic fibrosis.

Yes, I was very surprised to find out that I am a carrier of Cystic Fibrosis since we have no family history of the disease - and I should know since I am a genealogist! [20]

Two respondents were surprised at having an unexpectedly low risk for a condition. For one respondent this condition ran in her family. The other respondent unexpectedly found herself to be at a decreased risk of catching Norovirus.

6.5 Reason for Purchase

Respondents were asked their reasons for purchasing a DTC genetic test.[5] Interestingly, out of a total of 19 respondents, the largest group was five respondents who had purchased a test purely for genealogical reasons. Perhaps unsurprisingly, four of these respondents did not change their behaviour due to receiving their results, but one did. However, two of these respondents became interested in the health results as well.

> *Initially I tested only for genealogy however it has been very interesting seeing that my results mirror many of my ailments and so I follow the heath side as well. particularly via 23andMe*. [9]

Two respondents stated that they had purchased the tests mainly for genealogical reasons, but had also been interested in the health aspects. Interestingly both of these respondents had changed their behaviour after receiving their results.

> *I was primarily interested in genealogy but I also was interested in genetic traits, health and so on. I have an aunt who died of Lou Gehrig's disease, then found out two distant relatives on my mother's side had it so that was also a reason though not primary*. [35]

Four respondents stated that they had purchased the test for both health and genealogy reasons. One of these respondents described how she had been involved in genealogy for a long time, and had previously purchased an ancestry-only genetic test. She was considering purchasing her current test for a while, but then became interested in the health aspects as well as the ancestry after she developed a melanoma, and so decided to purchase one. Another respondent described how she was adopted and so did not know any of her family's health or genealogical information, which led her to purchasing a joint test.

Only one respondent stated that he had purchased the test for health reasons alone.

my reasons for buying the test were to identify rick factors and try to mitigate them [26].

[5] As stated in Research Methodology, potential respondents were asked to complete the survey (the source of the email addresses for the interviews) if they had purchased or were thinking of purchasing a DTC genetic test which included risk estimates for at least two health conditions. Therefore not all respondents had purchased a test for health reasons.

One respondent described how he was a medical student, and so purchased a test to be prepared for patients bringing results in to show him.

The other six respondents gave general reasons such as curiosity, novelty and wanting to find out what could be done with the tests. One respondent stated:

> *I bought the test because in general I am interested in science and tend to, um "geek out" about these kinds of things. Perhaps some vanity as well.* [8]

6.6 Information

6.6.1 Information seeking

Twenty-one respondents were asked questions about their information-seeking behaviour prior to purchase of a DTC genetic test. Sixteen of the participants stated that they had indeed searched for information. However, five of the respondents stated that they had not searched for information before purchasing a test.

6.6.1.1 Respondents who Sought Information

The sixteen respondents who stated that they sought information before purchasing a test varied on a continuum from those who generally read up about DTC genetic tests to those who only sought specific pieces of information.

At one end of the scale was one respondent who only searched for general information about DTC genetic tests. This respondent stated that he just generally read about the tests, mainly on science blogs. At the other end of the scale were two respondents who researched very specific details about the tests, but did not search for general information. Details searched for included the number of causal genetic markers tested, the interpretation of the data in relation to ethnicity and the genetics of specific diseases. As one of the respondents put it "*my research was mainly to understand the technical details of what I would be getting".* [14]

Six of the respondents searched for both general and specific information. For example, one respondent searched for "*how many genetic markers were typed, what tools they provided to interpret the data, and generally how useful the product is".* [15]

Another respondent searched for the error rate, the cost of the tests and the number of SNPs tested, as well as the general methods and the state of the field.

The remaining six respondents searched for general information but in certain areas. For example,

one respondent searched for the information which the company would provide, how the company used consumers' information and the effect that that would have on her. In contrast, another respondent was not concerned about the privacy issues as long as a privacy policy was provided, but searched for information relating to the capabilities and limitations of the tests, as well as testimonials from consumers.

Ten respondents described how the information that they had found influenced their decision to purchase a test. One simply stated that the information that she had found obviously influenced her. Two respondents described how the information that they had found convinced them to purchase a test. One of these respondents stated that it was the cost-effectiveness of the test he chose that convinced him. The other described how he found the markers that the company used, was familiar with them, and saw how they presented the results i.e. with the interpretation of the results using the markers. He stated that this information:

> *...persuaded **[me]** that 1. their price for the running my DNA against those markers was a fair price, and 2. they did a good job of presenting the data. So it was those two specific pieces of information that convinced me.* [15]

Two respondents stated that there was information that they had found without which they would not have purchased a test. For the first respondent this was information on the coverage of the test: both SNPs tested and the accessibility of the raw data. The second respondent stated that, although there was no one piece of information that it was necessary for him to find in order to purchase a test, if he had not found information on the wide range of topics for which he searched, such as the capability of the test, what could be done with it and information about the company, he would not have purchased it.

Five respondents described how the information that they found influenced their decision on which test to purchase. The first stated that, for the first test she purchased, it was the privacy policy of a particular company that convinced her to purchase from them. The second respondent stated that the selling point for the company she chose was their reputation for customer service. The third respondent stated that it was the information about the capabilities of the different tests that convinced her, namely that the health information provided by her company of choice would be useful in her situation. The fourth respondent stated that there were three groups of information that persuaded him to use 23andme:

> *1 - DNAForums comments about 23andMe*
>
> *2 - the fact that 23andMe gave dual results (medical & ancestry)*

3 - the fact that 23andMe gave more refined DNA haplogroup results at the time. [9]

However, although the information that he found persuaded him to use 23andme, he originally decided to purchase a test because of watching a documentary about the Genographic Project. The fifth respondent described how he had already decided to purchase a test before searching for information. However, discovering that 23andme offered free updates, access to raw data and good security policies convinced him to purchase a test from them. The fifth respondent described a thorough search for information about DTC genetic tests. However, she stated that most of this information did not influence her with regard to purchasing a test. The only two pieces of information that did influence her were the cost (when she noticed that it had decreased) and the number of diseases and conditions that were tested.

Two respondents described how they were not influenced by the information that they had found, as they had already decided to purchase a test prior to their research. For example, the first respondent described a large number of specific details that he had researched about DTC genetic tests, and stated that he had attended a presentation on them; however when asked if the information he had found influenced him replied:

> *No, I was pretty convinced by looking at other peoples results. My research was mainly to understand the technical details of what I would be getting.* [14]

The other respondent stated he knew that he wanted to purchase a test before conducting any research. However, although the information that he found did not influence him in this regard, he was unsure of some aspects of the tests before searching and so was reassured by what he found.

6.6.1.2 Respondents who did not Seek Information

In total, five out of the 21 respondents stated that they did not search for information about DTC genetic tests in preparation for their purchase.

Two respondents had purchased a DTC genetic test previously. Both had searched for information before purchasing their first test, and so already had a background knowledge of the area. However, the first tests purchased by both respondents were only focused on genealogy; therefore they would not have searched for health information. One of the respondents did report that she had previously read 23andme's website and set up a demo account; however this was not in preparation for purchasing a test. She had also accessed another individual's 23andme account and so would have been exposed to some of the information. The other respondent described a long use of

genetic tests, dating back to 2001, and generally keeping up with what the different companies were offering. She summed it up in the phrase "t*o stay informed I take the different tests".* [2]

The other three respondents had not previously purchased a DTC genetic test. One stated that he had been involved in a work project involving genomics, and so had researched genetic tests as part of that. He had mainly searched for information about the genetic variants using the scientific literature, and as an individual working in the field could be considered to be well informed. The second respondent, when asked if she had searched for information about the tests, replied "*Ho Ho No I just dove right in".* [4] However, she did mention having read genetic genealogy blogs in the past, which would have given some information about the tests; although health information would not have been included. She also demonstrated knowledge of the area:

> *The buzz at the time I tested years ago, was that a much larger portion of the Y chromosome would be tested. The amatuer geneticists were hoping to find SNP's unavailable through FTDNA and others at that time. These tests did result in more SNP discoveries.* [4]

The final respondent stated that she had not researched the test before purchase, but had relied on her daughter's judgement. However, she stated that her daughter was intelligent and had researched the area herself.

6.6.2 Responsibility

Respondents were asked whether it was the provider's or consumer's responsibility to ensure that consumers were fully informed about the tests.

By far the biggest group of respondents, 10 in total, stated that they thought it was both the provider's and the consumer's responsibility. Many of the respondents described how the provider should provide information, but that the consumer should read and understand it.

> *I think it's the company's responsibility to provide information but I think it's the customer's responsibility to read that information before buying a test, and to ask questions if there's anything they don't understand.* [17]

Three respondents placed the responsibility solely on the consumer.

> *Ultimately, the individual is responsible, and if they need a babysitter, then they shouldn't buy the product.* [2]

Conversely, one respondent stated that it was the provider's responsibility.

> *I strongly feel that the company has to be the one responsible because much of what one needs to know to understand these things requires college to have come into contact with them prior to testing. And even with that education level, it's impossible to know what the company is doing to process the information unless they tell us.* [33]

Two respondents also mentioned the government or regulators. One of these thought that both the provider and regulators should have responsibility. The other respondent thought that the responsibility should be the government's, with companies simply required to comply with regulation.

When asked which responsibilities providers have, the most common answers were to provide information and education about the tests to consumers.

> *On the question of responsibility, I think the company has a responsibility to provide "all important information" on subjects like coverage, privacy, etc.* [23]

Other responsibilities mentioned were accuracy, truth, completeness of information, ease of access of information and to suggest further information if the consumer desires. One respondent thought that providers should aim to fully inform consumers, and another that they should be clear about the ambiguity of any interpretations. One respondent stated that providers should inform consumers about the complexity of the results, and how they may evolve over time. Another suggested responsibility was that providers should inform consumers if anything changes.

When asked what responsibilities consumers have, the most common answers were to read and understand the information provided by the provider.

> *I think it's the company's responsibility to provide information but I think it's the customer's responsibility to read that information before buying a test.* [17]

Other responsibilities mentioned were to ask questions if needed, to learn about the tests and be knowledgably and fully informed, to educate themselves and to understand what they are paying for and what they are 'getting into'.

6.6.3 Information Provision

Respondents were asked what information about DTC genetic tests they thought should be provided by the companies that sell the tests.

A wide variety of different answers were given. The most common answers, given by six respondents, related to what the product is. This included the coverage of the tests, the technical details, what the product does and simply what the tests are.

The next most common answers related to how the tests work, what they involve and methods.

Three respondents thought that providers should provide information on the tests' uses and limitations, and what can be done with them.

> *If testing companies are very clear about what their product is and its uses/limitations, then the consumer should have no grounds for complaint*. [19]

Two respondents thought that providers should provide information on how to interpret data, and two more thought that there should be information on the basics.

All other answers were given by one respondent apiece, and included: all of the information that is obtained through sequencing, the possibility that results might change in the future, the conclusions that can be drawn from the sources used by the company to base their analyses, contact details, a recommendation to contact them for further information, a demo, the evidence on which analyses are based, FAQs, future technologies, facilities for further analysis, information about gene-gene-environment interactions, information about genetic architecture, information about government regulation, how information will be used, information about each SNP, information about the relevance of results, medical implications, ownership of data, information about privacy and information about statistics.

6.6.4 Rating of Information Provision

Respondents were asked to rate the provision of information on the website of the provider from which they purchased a test.

The largest group of respondents, six in total, rated the information provision as good.

> *For anyone reasonably literate, enough simple info is given, and it is easy to understand. Having other references for the more interested/curious is also good*. [12]
>
> *At least with 23andMe, their website provides information to help you understand what your results means and can give you a basic understanding of what you need to know. So at least with this one company I would say you could get by with little upfront knowledge because more will be provided for you as you get your results.*[8]

Most of these respondents thought that the website contained enough information for consumers to make a decision without the need for other sources.

> *I certainly think the 23andme webpage has enough information for most people*. [3]

Four respondents thought that the website was good, but was lacking something. For example, one respondent thought that the website should have a better way of presenting the results and helping people to interpret them. Another thought that there was not enough information about the evidence on which new results are based, and about what would happen to the DNA sample with regard to further tests. A third respondent thought that the information provided was good, but that it would be a good idea for consumers also to use other sources. The fourth respondent thought that most of the information was good, but disliked how the privacy terms on one website were changed after she had purchased a test.

Three respondents described how they had used more than one provider, and that the websites were very different in terms of information provision. One respondent described how one website he had used had had a lot of easy-to-access information, another did not have much (it was based on a low-price model) and a third had some but it was difficult to use. Another described how the information provided varied over the years and between providers:

> *The websites have changed over the years, and luckily those businesses that promised more than they could deliver are mostly out of business. But of those companies left, there are different levels of information/tutorials available.* [19]

The third respondent stated that consumers should definitely use other sources of information:

> *other sources needed for sure. the websites of the company I bought the test as well as others I checked we're to full of fancy pictures but scarce usefull information. exception made for some companies blogs.* [18]

6.6.5 Sources

Respondents were asked what sources of information they thought were suitable for consumers to use to research DTC genetic tests.

Answers covered a large variety of different sources. The most common answer (with six citations) was blogs; either blogs in general, science blogs, or specific blogs. For example, one respondent stated: "*various blogs (gene sherpa, genomes unzipped, etc)".* [18] Another stated:

> *I also think simple Google searches are useful as they lead to blogs of people not affiliated with the companies providing reviews of the services*. [3]

The joint second most common answer (with four citations) was Wikipedia.

> *My experience with Wikipedia, is that as long it's not something that can touch in on national pride, ex. History, language etc, as well as race or religious/spiritual belief, then one is on relatively safe ground on Wikipedia.* [13]

Joint second with Wikipedia was mailing lists and forums, with 23andme's forum one of those mentioned. One suggested mailing list was that of the ISOGG, an organisation mentioned by three respondents in total.

Other sources mentioned included scientific publications (mentioned by two) and a wide variety of sources mentioned by one respondent each: a community website named 23andyou, articles, books, citizen science sites, general surfing, the government, National Institute of Health, National Center for Biotechnology Information public information sites, health professionals, Lifehacker, the PhG foundation, reviewing organisations, seqanswers.com, snpedia and the providers' website.

6.6.6 Kept Up With Area

Respondents were asked if they had kept up with the area of DTC genetic tests after receiving their results.

Most respondents (11) stated that they continued to keep up with the area. Three respondents mentioned that they keep up with what tests are available, with one stating that he also reads blogs about it. One respondent described how he sometimes follows links posted on 23andme's forum, and that new results are always being provided which he often researches. Another respondent stated that he looks into 23andme's new tests when they email about them. A third respondent stated:

> *I do keep up, I think it is a very important issue for the future in terms of public awareness of science and genetics in general. Plus, I want to upgrade my personal results if something better comes along.* [14]

One respondent described how she checks the sites daily, but mainly for genealogical reasons. Another respondent stated that he discusses the tests in his medicine classes and has written an article on them. Finally, one respondent (who writes a blog) described how she takes tests, reads

papers, writes articles, reads mailing lists, talks with management and communicates with the people in her DNA project in order to keep updated.

6.7 Understanding

Respondents were asked how easy they thought the genetic test they used was to understand.

Most respondents, 19 in total, thought that it was easy or relatively easy to understand, or stated that they had had no problems using or understanding it.

> *They were very easy to understand. 23andMe has two types of information: layman's info and technical info. They do a good job explaining everything at a level most people can understand, and they also have another page providing technical information, so people with a college level or higher knowledge of genetics can learn more.* [27]

It should be noted that some respondents added the caveat that their backgrounds (e.g. worked in a medically-related job) meant that they may have found the test easier to understand than the general public.

Two respondents thought that the tests may not be easy for the general public to understand. Although they themselves had had no problems (due to their experience in the area), they thought that the public might do, especially in understanding higher and lower risks.

> *... it could be slightly more confusing, particularly for people who've just started using the service. The overall plus-and-minus summary is well done, but accepting it blindly could be misleading.* [1]

Three respondents stated that the tests had either been difficult to understand at first or that they had had to look up a lot of the information, but that they had managed to understand it eventually.

> *I had to look up nearly every term I came across, many acronyms and biological system relationships. 23andMe decodes what particular flags are for, and includes information on the relevance, which is helpful.* [11]

The remaining two respondents stated that they had understood some of the results, but not understood them completely. One respondent had not entirely understood how all of the different risk variables interact, and the other respondent was unsure which populations the results were relevant for.

Respondents highlighted some of the parts of the tests or results that they thought were more difficult to understand. Difficulties mentioned (by one respondent each) were difficulty in finding for which populations the results were relevant, understanding the DNA sequencing, understanding the process by which the results were obtained and understanding the forums on the provider's website. As described above, one respondent reported the need to look up a lot of the terms and other information on the website. Another respondent reported unfamiliarity with many of the terms. One respondent mentioned that, since risk factors were based on American populations, she was unsure how that related to UK populations.

> *For example, my highest risk factor was for obesity (49% versus the average risk of 59%), but the map they showed was for obesity statistics in America and nearly all the papers they cited were based on studies done on American populations. All I could really conclude from this was that I had some genes that might make me somewhat less disposed to obesity than the average American.* [17]

The same respondent mentioned that it was misleading that some increased disease risks were shown in red, even though they were insignificant in real terms. A respondent whose first language was not English mentioned that he had had to translate some of the medical terms, but that he had not found this too difficult to do using Wikipedia. Finally, one respondent mentioned that the raw data was difficult to understand.

Two information topics were highlighted as important topics for consumers to understand. The first was overall risk, mentioned by two respondents.

> *I find them fairly easy to understand, but I realize close attention must be paid to the overall risk. Sometimes, a person may have an increased risk for a disease, but the overall risk is still very low.* [20]

The second was the importance of consumers understanding their environmental risk along with their health risk in order to fully understand their susceptibilities; something which is not always clear.

> *The 'environmental' risk is just as important for the novice to see as the 'genetic' risk. The environmental risk is given elsewhere in the genetic health report, 'hidden' on the next page from the genetic results. Consequently, some people might not appreciate that risk and instead freak out over genetic health risk unnecessarily.* [12]

Three interesting points with regard to the ease of understanding the information provided were

mentioned by respondents. Firstly, that there was helpful information provided on providers' websites. Secondly, that more health-related information was provided on the websites than would be provided by a doctor. Thirdly, that both simple and complex information is provided so that consumers can read to their level of understanding.

Three tools to help consumers understand their results were mentioned by respondents. These were Wikipedia, 23andme's forums and the user tutorials.

6.8 Genetic Knowledge

Respondents were asked if it was necessary for consumers to have a basic level of genetic knowledge in order to understand the tests. Three respondents stated that they thought consumers needed a basic level of genetic knowledge.

> *Yes, if consumers aren't at a basic level of education - like a university intro to genetics course - they may have wild ideas about how genetics work. There are plenty of myths.* [11]

Two respondents thought that only a very basic level of genetics was required.

> *Most and perhaps all of the basic knowledge can be found just by looking it up on wikipedia, and large parts of it are basic teachings in school.*
>
> *I would claim that most of the things are presented in such an easy to understand way, on 23andme, that hardly more than the above are needed.* [13]

One respondent stated that a basic knowledge of medical terms and statistics would be useful in helping people to understand the results. Two respondents thought that only knowledge of statistics was required when examining the disease risk information.

> *As for risk testing... I think it would be better for people to understand statistics than genetics before reading those reports :)* [1].

Another respondent thought that a basic knowledge of genetics was important but not necessary, one thought that it depended on which website was used and a third stated that it was up to the individual consumer.

Two respondents stated that a basic genetic knowledge was not necessary, at least not to begin with.

> *No, just as it is not necessary to know how an internal combustion engine works before buying a car. (Of course, the additional knowledge can come in handy in both cases.)* [15].

Respondents mentioned pieces of information that they thought it important for consumers to know. These included: genes are not entirely responsible for phenotype (i.e. there are also environmental influences), results cannot simply be viewed as either a negative or positive dichotomy, humans have two chromosomes and alleles can be dominant or recessive.

Several respondents mentioned that the level of genetic knowledge necessary to make full use of ancestry genetic testing is much higher than for health testing.

6.9 Other or Anything to Add

6.9.1 Other

Four respondents gave answers that could not be grouped into any of the previous topics, but were nevertheless interesting.

One respondent mentioned the technology he would like to see available in the future. He stated:

> *I hope that data mining / machine learning continue to build inferences between genotypes and phenotypes. I want to be binned into a class that generalizes humans like myself and could give instructions on how to eat, exercise and study the best for a long and productive life.* [11]

This respondent also mentioned that he would like to eventually have his entire genome sequence, and to develop open-source tools that would help users find out information about their genome.

The second respondent stated that he thought that in the future it would be possible to alter your genome and repair tissues with stem cells. He stated:

> *I might be able to fix both my back and the osteoarthritis via stuff like stem cells from my own body. Within the next 20-40 years rejuvenation might be possible, and with my curiosity I want to see as much of the future as I can ;-)* [13]

The third respondent mentioned how useful genetic health information can be, and that although it does not tell you exactly what will happen it can save lives. She compared worries about consumers having access to genetic testing with those that doctors had when consumers could first buy pregnancy testing kits over the counter (i.e. flawed arguments about protecting the consumer), and

believes that it is about money rather than safety concerns. She was adamant that although genetic testing is far from a mature technology individuals have the right to know information about their own DNA, and that patients can often be more knowledgeable about the fine detail of their health than doctors. She expressed the hope the genetic testing will become more common. On a different topic, she stated that the law must protect consumers from having their information used by companies without their explicit position.

The fourth respondent was very opposed to the regulation of DTC genetic testing companies. She thought that there should be some laws that apply to all companies, such as truthful advertising, but that regulation would "*kill off this new industry*". She stated:

> *My question, is why do you need regulation by industry, which invites rent seekers to lobby? For example, those that want to require a doctors perscription to get a test, and those that want to require genetic counseling. It is ALL ABOUT MONEY, dressed up as protecting us poor stupid people from ourselves.* [2]

6.9.2 Anything to Add

All respondents who completed the interviews were asked if there was anything about genetic tests that they wished to add, whether or not it was related to the questions asked during the interview. Although the majority of respondents did not have anything to add, 10 of them did.

The first respondent stated that he believed that genetic testing will start to become much more common for parents to purchase for their unborn children; although self-testing will become more common, it will take longer to do so than testing during pregnancy. He also predicted that once full genome sequencing becomes cheap enough, people will only ever need to buy one actual sequencing test (which may be bought for them by their parents before they are born) and that the market will start to move towards interpretation of the results.

The second respondent stated that, due to the changes genetic testing made to her life, she has had most of her family tested. She stated:

> *i [sic] believe this technology is the future of individualized medicine , i can now trace on paper which traits, disease, risk my youngest child is at risk for because both sides parents have tested*. [10]

Once her granddaughter has been tested then three generations of her family will have done so, and she hopes that babies will soon be tested at birth.

The third respondent stated:

> *Genetic tests aren't as nearly as useful as they can be, but they are presently useful enough for the cost. The usefulness is only increasing with time.* [11]

The fourth respondent stated that he was glad that this research (i.e. the interviews) was being done, and that a better public understanding of genetics would be good. He also mentioned the worry that DTC genetic tests may be regulated so that the public cannot access them themselves. He stated:

> *The practical utility of genome sequencing for medical genetics may turn out to be limited for the public since most people will not carry a simple Mendelian trait of interest but for personal ancestory and the odd rare de novo mutation, everyone should have the right to their information.* [14]

The fifth respondent described her strong belief in open access to information, and that the patient is the one who should be acting on their own information. She believes that the results of medical genetic testing have the ability to save lives, especially for conditions that are much easier to treat if caught early, and that carrier and drug sensitivity testing are also very important. She also mentioned ancestry testing, and stated that discovering information about your ancestry can bring people closer together.

The sixth respondent stated:

> *The only think I would like to add about genetic tests is that I think everyone should have the right to access their own genetic data and not be forced to order a test through a doctor or a genetic counsellor.* [17]

The seventh respondent described how, in the country in which he lives, doctors are generally ignoring DTC genetic tests. He adds that doctors are not opposed to the tests in themselves, and that their attitude is slowly changing.

The eighth respondent stated that he hopes that DTC genetic tests start to be used more and that when people think about DNA testing they will think of them, rather than just thinking of criminal and paternity testing.

The ninth respondent stated that he hopes that genetics can be considered to be the same as other health information, and that it is better understood by professionals. He stated that:

> *The main danger I see at the moment is that the majority of commercial offerings are of low or no value, either through ignorance of the creators or through blatant exploitation, and the only way to get rid of these is through information and education.* [31]

Lastly, the tenth respondent stated that she has had a lot of fun with the ancestry side of genetic testing. She also described how the carrier testing had been useful for her family, and that the health information was good to have. She stated:

> *I would say that genetic testing has been one of the most interesting things!* [7]

6.10 Summary

The results and analysis of the email interviews allowed for an in-depth examination of many issues relating to DTC genetic tests.

As described in section 5, the survey results indicated a minority of consumers changed their health behaviour due to the information generated by the tests. The email interviews confirmed these findings, with 17 respondents describing specific changes that they had made after receiving their results. Several reasons for these changes were identified, including a high risk for a specific disease, a comparison of results with family history and a general motivational effect of receiving the results. Reasons were also identified for respondents who had not changed their behaviour, such as no cause for concern in their results, knowledge of their family history and existing good health behaviour.

The email interviews also helped to clarify the survey's mixed findings for changes to consumers' health anxiety after receipt of the information generated by the tests (see section 5), with 13 respondents stating that their health anxiety did indeed change due to receiving their results. Most had noticed a decrease in health anxiety, with many stating that this had been sustained. However, two did notice both a decrease and an increase. Reasons for a decrease in health anxiety included a lack of an increased risk for diseases for which the respondent had a family history and simply knowing the conditions for which they had an increased risk. Reasons for an increase in health anxiety included an increased risk for certain conditions and thinking about health more frequently after receiving their results. Reasons were also identified for why some respondents did not notice a change in health anxiety, including respondents' attitudes towards health, awareness that genetics is one of many disease variables and a lack of worrying results.

Respondents described their information-seeking behaviour and opinions about the information provided by the company from which they purchased a DTC genetic test. Most respondents stated that they had sought information before purchasing the test, with all but one of those who had not searched for information describing some background knowledge of the area. Some respondents read generally about the tests and related area, whereas others sought specific pieces of information, sought for both general and specific information or read generally in specific areas. Most respondents thought that both consumers and providers shared responsibility for consumers to be fully informed about the tests. In contrast, some thought that it was consumers' responsibility alone, one that it was solely the providers' and two that government or regulators should have a role. When asked what information about the tests companies should provide, respondents gave a variety of answers including information about the coverage of the tests, their uses and limitations and how the risk information is used. Most respondents thought that the information provision on the website from which they purchased a test was good, although several of these thought that they were lacking something, and one respondent stated specifically that consumers should use other sources of information. Respondents thought that many different sources of information were suitable for consumers to use, from blogs and mailing lists to Wikipedia and scientific publications.

Many opinions about the tests and related area were also identified, along with respondents' experiences with the tests. For example, respondents purchased a test for a variety of reasons, from health to genealogy to general interest. Most thought that the results were easy to understand, although a small number disagreed. Almost two thirds of respondents had shared their results with a health professional, and only one respondent was against the idea in principle. Out of those who had shared their results, most had received a positive response, although some had received a negative or indifferent one. A minority of respondents were surprised by parts of their results, but well over half were not.

This study has produced a large amount of useful data, much of which is unique in the literature.

7 Content Analysis

This chapter presents and analyses the results of the content analysis and discusses the findings. It is split into two main sections: Results (section 7.1) and Analysis and Discussion of Content Analysis Results (section 7.2).

7.1 Results

This section presents the results of the content analysis for each group of items. Items coloured green were those derived from consumers' survey answers, items coloured red were those derived from professional recommendations and items coloured blue were those jointly derived from both.

7.1.1 Accuracy

This section included all 28 items that related to the accuracy of the tests, accuracy of the analysis, evidence for the analysis or value of the analysis (see Table 22).

The results showed that 23andme covered the most items, 24 in total. The only items not covered by 23andme were specificity of the test, variability in accuracy, confidence intervals of results and risk of contamination. However, three of these four were not covered by any of the other websites and variability in accuracy was only covered by five. The website which covered the least items was Testcountry, which only covered two: predictive value of the test and diagnostic value of the test. Two items were covered by all 13 websites: predictive value of the test and diagnostic value of the test. Three items were not covered by any of the websites: specificity of the test, confidence intervals of results and risk of contamination.

Sixteen of the items were derived from the recommendations of professional associations. These included one of the items covered by all of the websites, predictive value of the test, and one of the items not covered by any website: specificity of the test. Sixteen of the items were derived from consumers' survey answers. These included one of the items covered by all of the websites, diagnostic value of the test and the three items not covered by any of the websites: specificity of the test, confidence intervals of results and risk of contamination.

The mean number of websites per item was 6.2. For those items derived from the recommendations of professional associations the mean was 6.6 and for those items derived from consumers' survey results the mean was 5.7.

Table 22 Coverage of Items in Accuracy Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
General accuracy of the test	✓		✓		✓			✓	✓	✓	✓	✓		8
Accuracy of sequencing	✓								✓	✓	✓			4
Sensitivity of the test	✓								✓	✓				3
Specificity of the test														0
Reliability/repeatability of the test/results	✓		✓						✓	✓				4
Predictive value of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Populations for which the information is known	✓	✓	✓									✓		4
Scientific evidence available for the population	✓		✓	✓							N/A			3
Accuracy of the interpretation of the results/predictions	✓		✓	✓		✓	✓	✓	✓		✓			8
General scientific credibility of the results	✓		✓	✓	✓	✓	✓	✓	✓		✓	✓		10
Difficulties with establishing clinical validity	✓	✓	✓	✓	✓	✓	✓	✓	✓		✓			10
Evidence for interpretations of the results	✓		✓		✓	✓		✓	✓		✓	✓		8
(If evidence is presented) Reference to the criteria used to include and/or exclude published literature	✓		✓	N/A			N/A		✓		✓			4
Evidence/links to evidence of the association between a genetic marker and a disease, condition or trait	✓		✓		✓			✓	✓		✓	✓		7
Information about the association between a genetic variant and a disease, condition or trait.	✓	✓			✓	✓	✓			✓	✓			7
Analytical validity of markers	✓		✓			✓		✓			✓			5
Clinical validity of markers	✓	✓	✓		✓	✓	✓	✓	✓		✓			9
General validity of tests/science	✓	✓	✓	✓		✓	✓	✓	✓		✓	✓		10
Quality of data	✓		✓				✓	✓	✓	✓	✓	✓		8
Quality of/confidence in analysis	✓		✓	✓	✓		✓	✓	✓		✓			8
Error rate	✓								✓	✓				3
Variability in accuracy			✓	✓	✓			✓			✓			5
Outside verification of accuracy	✓							✓	✓					3
Confidence intervals of results														0
Diagnostic value of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Adequacy of the interpretation of the results	✓	✓	✓	✓	✓		✓	✓	✓		✓	✓		10
Risk of contamination														0
Changeability of current research	✓	✓	✓	✓				✓	✓		✓			7
Total	**24**	**9**	**20**	**11**	**12**	**10**	**11**	**17**	**20**	**9**	**19**	**10**	**2**	**174**

7.1.2 Vendor

This section included all 18 items that related to the vendor or to the laboratory in which the testing was conducted (see Table 23).

Table 23 Coverage of Items in Vendor Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
CLIA certification of laboratory or other accreditation	✓		✓	✓			✓	✓	✓		✓	✓	✓	9
Details of Laboratory			✓				✓		✓			✓		4
Operator of services	✓		✓				✓		✓			✓		5
Location of operator			✓				✓							2
Funding arrangements	✓		✓						✓		✓			4
Advertising arrangements	✓										✓			2
Use of established testing procedures by laboratory	✓		✓	✓	✓		✓	✓	✓		✓	✓		9
Details of company	✓	✓	✓		✓				✓	✓	✓	✓		8
Company's years in business	✓										✓	✓	✓	4
Ownership of company	✓		✓								✓			3
Publishing records of company scientists			✓											1
Long term commitment of company	✓		✓		✓				✓		✓	✓		6
Provision of answers to consumer questions	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Individuals associated with the company	✓		✓		✓	✓			✓		✓	✓		7
Support before testing	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Support after testing			✓	✓	✓	✓	✓				✓	✓		7
Support for individuals			✓	✓		✓	✓			✓	✓	✓		7
Support for families														0
Total	**12**	**3**	**15**	**6**	**7**	**5**	**9**	**4**	**10**	**4**	**13**	**12**	**4**	**104**

The results showed that DeCODEme covered the most items, 15 in total. The only items that it did not cover were advertising arrangements, company's years in business and support for families. However, support for families was not covered by any of the websites, advertising arrangements by only two of the websites and years in business by only four of the websites. Accumetrics/Viaguard only covered three items, the least items for any website. These were: details of company, provision of answers to consumer questions and support before testing. Two items were covered by all of the websites: provision of answers to consumer questions and support before testing. Only one item, support for families, was not covered by any of the websites.

Six of the items were derived from the recommendations of professional associations. These included one of the items covered by nine websites, CLIA certification of laboratory or other accreditation, and the two items that were only covered by two of the websites: location of operator and advertising arrangements. Fourteen of the items were derived from consumers' survey results. These included both of the items that were covered by all of the websites: provision of answers to consumer questions and support before testing, and the two least-covered items: support for families (covered by no websites) and publishing records of company scientists (covered by one website).

The mean number of websites per item was 5.8. For those items derived from the recommendations of professional associations the mean was 4.3, and for those derived from consumers' survey results the mean was 6.5.

7.1.3 Disease

This section included all nine items that related to disease or interventions (see Table 24). Two of the items are not included in the written description or statistics as they were not applicable to several websites (see footnote underneath Table 24).

The results showed that DeCODEme covered the most items, six in total. The only item not covered by deCODEme was percentage with disease whose disease expression had a strong genetic component. However, only two other websites covered this item. EasyDNA and International Biosciences both covered none of the items. The item covered by the highest number of websites was recommendations for appropriate actions based on results, which was covered by 11 items. The item covered by the fewest number of websites was percentage with disease whose disease expression had a strong genetic component, which was only covered by two.

Five items were derived solely from the recommendations of professional groups. These included both the three most-covered and the two least-covered items. Only two items, possible prevention strategies and actionability of results, were derived from consumers' survey results. These had a middling coverage by the websites, seven and eight respectively.

The mean number of websites per item was 7.1. For those items derived from the recommendations of professional associations the mean was 7, and for those derived from consumers' survey results the mean was 7.5.

Table 24 Coverage of Items in Disease Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Population at risk for disease		✓	✓								✓	✓		4
Percentage with disease whose disease expression had a strong genetic component		✓								✓				2
Behaviour that influences development of a condition	✓	✓	✓		✓	✓			✓	✓	✓	✓		9
Environmental factors that will influence development of a condition.	✓	✓	✓		✓	✓			✓	✓	✓	✓		9
Evidence for recommended interventions±			✓				N/A	N/A					N/A	1
Evidence against recommended interventions±							N/A	N/A					N/A	0
Recommendations for appropriate actions based on results	✓	✓	✓		✓	✓	✓		✓	✓	✓	✓	✓	11
Possible prevention strategies	✓		✓		✓	✓			✓	✓	✓			7
Actionability of results	✓	✓	✓		✓	✓	✓		✓		✓			8
Total	**5**	**6**	**7**	**0**	**5**	**5**	**2**	**0**	**5**	**5**	**6**	**4**	**1**	**51**

7.1.4 Benefits

This section included all 13 items that related to the benefits or usefulness of the tests (see Table 25). One of the items is not included in the written description or statistics as it was not assessed for most of the websites (see footnote underneath Table 25).

23andMe, Mapmygene, Navigenics and Pathway Genomics covered the joint most items, 11 in total. The only item not covered by 23andMe, Mapmygene and Navigenics was evidence for benefits. The only item not covered by Pathway Genomics was immediate usefulness of the test. EasyDNA, International Biosciences and Testcountry covered the joint least number of websites, eight in total. Eight of the items were covered by all of the websites. The item covered by the least number of websites was evidence for benefits, which was only covered by one.

Seven of the items were derived from the recommendations of the professional associations. These included five of the items covered by all of the websites. They also included evidence for benefits, the item covered by only one of the websites. Five of the items were derived from consumers' survey responses. These included three of the items covered by all of the websites: general

± These items are not included in the written description or statistics as they are not applicable to several websites.

Table 25 Coverage of Items in Benefits Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Benefits of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
Evidence for benefits												✓		**1**
Clinical usefulness of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
Scientific usefulness of the test	✓								✓	✓	✓	✓		**5**
General usefulness of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
Immediate usefulness of the test	✓		✓		✓		✓			✓	✓			**6**
Ability of results to help inform health behaviour choices	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
Process by which results can help inform health behaviour choices	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
Informative value of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
No overstatement of utility[±]	N/A	N/A	N/A	N/A	N/A	N/A	N/A	✓	✓		✓	N/A	N/A	**3**
Process by which information can be used	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
Information that can be learnt from the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	**13**
Specific applications of the results	✓	✓	✓		✓	✓	✓		✓	✓	✓	✓		**10**
Total	**11**	**9**	**10**	**8**	**10**	**9**	**10**	**9**	**11**	**11**	**12**	**11**	**8**	**129**

usefulness of the test, process by which information can be used and information that can be learnt from the test.

The mean number of websites per item was 10.5. For those items derived from the recommendations of professional associations the mean was 10.1, and for those derived from consumers' survey results the mean was 11.0.

7.1.5 Limitations

This section included all 21 items that related to the limitations, risks of the tests or risks of the treatment/investigation (see Table 26).

The results showed that 23andme covered the most items, 17 in total. The only items that it did not cover were risks of the test with regard to family members, possible implications or consequences for family members, consequences of investigation and consequences of treatment. However, risks of the test with regard to family members and possible implications or consequences for family members were only covered by two websites each, and consequences of investigation and

[±] This item is not included in the written description or statistics as it was not assessed for the majority of websites (see methods).

Table 26 Coverage of Items in Limitations Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
General limitations of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓		✓			10
Limitations of the test with regard to claimed benefits	✓		✓	✓		✓		✓	✓	✓	✓	✓	✓	10
Limits to informative value	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Limitations of the interpretation of results	✓	✓	✓	✓		✓	✓	✓	✓		✓	✓		10
Possible dangers	✓					✓			✓		✓			4
General risks of the test	✓								✓		✓			3
Psychological risks of the test	✓		✓			✓		✓	✓		✓		✓	7
Risks of the test to family members						✓				✓				2
Risks of the test with regard to general insurance discrimination	✓		✓							✓	✓	✓		5
Risks of the test with regard to employment discrimination	✓		✓							✓	✓	✓		5
Risks of the test with regard to life insurance discrimination	✓										✓			2
Risks of the test with regard to disability insurance discrimination	✓										✓			2
Risks of the test with regard to long-term care insurance discrimination	✓										✓			2
General risks of disclosure of information	✓					✓								2
Risks of disclosure of information to a web community	✓		✓			✓								3
Possible implications or consequences for consumer	✓		✓			✓					✓			4
Possible implications or consequences for family members						✓				✓				2
Consequences of investigation												✓		1
Consequences of treatment											✓			1
Downfalls of taking the test	✓					✓			✓		✓			4
Safety of the test	✓								✓	✓				3
Total	**17**	**3**	**9**	**4**	**2**	**12**	**3**	**5**	**9**	**7**	**15**	**6**	**3**	**95**

consequences of treatment were only covered by one website. The website that covered the least items was Geneplanet, which only covered two: general limitations of the test and limits to informative value. Only one of the items was covered by all of the websites: limits to informative value. Two of the items, consequences of investigation and consequences of treatment were only covered by one of the websites. Only three of the items were derived from consumers' responses.

These were all covered by low numbers of websites: risks of the test with regard to general insurance discrimination by five, downfalls of taking the test by four and safety of the test by three. All of the other items were derived from professional recommendations, including the item covered by all of the websites, limits to informative value, and the items covered by only one of the websites: consequences of investigation and consequences of treatment.

The mean number of websites per item was 4.5. For those items derived from the recommendations of professional associations the mean was 4.6, and for those derived from consumers' survey results the mean was 4.0.

7.1.6 Results

This section included all 27 items related to the results or how the results would be presented (see Table 27). Five of the items are not included in the written description or statistics as they were not applicable for all of the websites (see footnote underneath Table 27).

The results showed that DeCODEme covered the most items, 20 in total. The only items that it did not cover were possibility of finding out about conditions for which treatment is not available and statement of who interprets results (e.g. consumer or counsellor). However, possibility of finding out about conditions for which treatment is not available was only covered by five websites and statement of who interprets results (e.g. consumer or counsellor) by three. Inherent Health and Testcountry covered the joint least items, five in total. Meaning of results was the only item covered by all 13 websites. The item covered by the least number of websites was consumer access to analysis, which was only covered by one website.

In total, eight of the items were derived from professional recommendations. These included two of the items covered by 12 websites, highly nuanced nature of results and unsuitability of results to be used alone for medical decision making, and the item covered by three websites: statement of who interprets results (e.g. consumer or counsellor). Fifteen of the items were derived from consumers' survey answers. These included the only item covered by all of the websites, meaning of results, and the item covered by the least number of websites: consumer access to analysis.

The mean number of websites per item was 7.9. For those items derived from the recommendations of professional associations the mean was 8.3, and for those derived from consumers' survey results the mean was 7.6.

Table 27 Coverage of Items in Results Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Highly nuanced nature of results	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓		12
Possibility of finding out about conditions for which treatment is not available		✓		✓		✓			✓		✓			5
Possibility of finding serious health problems	✓	✓	✓	✓		✓			✓	✓	✓			8
Nature of the risk (either absolute or relative)	✓		✓	✓	✓	✓		✓	✓	✓	✓	✓		10
Unsuitability of results to be used alone for medical decision making	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	12
Manner in which test results are provided	✓		✓	✓	✓	✓		✓	✓	✓	✓			9
(If change in medicine recommended by results) Information about change in medicine provided on a link.[±]	✓	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	1
Provision of updated test results to consumers	✓		✓		✓				✓	✓	✓	✓		7
Recommendation that consumers should not alter medicine based on results but take to doctor[±]	✓	✓	✓	N/A	✓	✓	✓	✓	✓	✓	✓	✓	✓	12
Statement of when results can only give relative, rather than absolute, risk[±]			✓	N/A	✓	✓		✓	✓	✓	✓	✓		8
Ability to share results	✓		✓						✓			✓		4
Ability to match results with other users	✓		✓											2
Consumer access to raw data	✓		✓				✓		✓	✓	✓			6
Updates to interpretations	✓		✓		✓				✓	✓	✓	✓		7
Provision of help with regard to interpretation of updates[±]	✓		✓					N/A	✓		✓	✓		5
Description of user interface[±]	✓		✓	✓	✓			N/A	✓	✓	✓			7
Description of what consumers should expect with results	✓	✓	✓	✓	✓	✓		✓	✓	✓	✓		✓	11
Nature of data produced	✓		✓	✓	✓	✓		✓	✓	✓	✓	✓		10
Information provided	✓	✓	✓	✓	✓			✓	✓	✓	✓			9
Meaning of results	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Consumer access to analysis			✓											1
Detail of results	✓	✓	✓	✓	✓			✓	✓		✓		✓	9
Types of information or other areas that results can be compared to	✓		✓	✓	✓	✓		✓	✓		✓			8
Statement of who interprets results (e.g. consumer or counsellor)				✓							✓	✓		3

[±] These items are not included in the written description or statistics as they are not applicable to all of the websites

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Provision of help with regard to interpreting results	✓		✓	✓		✓	✓				✓	✓		7
Appropriate explanation of data provided	✓	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	12
Demo or sample version of results	✓		✓	✓	✓			✓	✓	✓	✓			8
Total	**23**	**10**	**24**	**17**	**17**	**13**	**6**	**14**	**22**	**17**	**23**	**14**	**6**	**206**

7.1.7 Ethical/Legal Issues

This section included all 15 items that related to legal or ethical issues (see Table 28). One of the items is not included in the written description or statistics as it was not applicable for all of the websites (see footnote underneath Table 28).

The results showed that 23andme covered the most items, but this was only seven, less than half of the total number of items. Accumetrics/Viaguard did not cover any of the items. None of the items was covered by all of the websites. The item that was covered by the highest number of websites was statement that third parties (e.g. law enforcement) may have access to samples if required by law, which was covered by 10 websites. Six of the items were not covered by any website.

All but one of the items was derived from the recommendations of professional associations. The only item derived from consumers' responses was situation with regard to health insurance, which was covered by six of the websites. The mean websites per item for the whole section was 2.3. The mean for the items derived from professional recommendations was 2.0.

7.1.8 Privacy and Security

This section included all 47 items that related to privacy issues, privacy policies, privacy controls, security issues, security procedures, confidentiality, data protection issues or similar (see Table 29). Six of the items are not included in the written description or statistics as they were not applicable for all of the websites (see footnote underneath Table 29).

The results showed that Pathway Genomics covered the most items, 34 in total. Accumetrics/Viaguard only covered one item, confidentiality of test, the least number of items for any website. No item was covered by all of the websites. However, four of the items were covered by 12 websites. These were: general privacy issues, general privacy policy, confidentiality of test and

Table 28 Coverage of Items in Ethical/Legal Issues group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Statement with regard to concerns over testing of children											✓			1
Statement that tests that do not meet clinical validity requirements should not be carried out in children[±]					N/A		N/A							0
Legal position of declaring results for insurance in general	✓						✓		✓		✓	✓		5
Legal position of declaring results for disability applications	✓													1
Legal position of declaring results for life insurance	✓								✓		✓			3
Legal position of declaring results for mortgage insurance														0
Legal position of declaring results for travel insurance														0
Possibility of change in legal position in future	✓													1
Statement that the results can be important for relatives	✓		✓							✓	✓			4
Statement that relatives have a right to know														0
Statement that relatives have a right not to know														0
Statement that third parties (e.g. law enforcement) may have access to samples if required by law	✓		✓	✓	✓		✓		✓	✓	✓	✓	✓	10
Statement that taking DNA from someone else is ethically inappropriate														0
Statement that taking DNA from someone else is a criminal offence in some jurisdictions								✓						1
Situation with regard to health insurance	✓		✓				✓		✓		✓	✓		6
Total	**7**	**0**	**3**	**1**	**1**	**0**	**3**	**1**	**4**	**2**	**6**	**3**	**1**	**32**

data protection issues. The items covered by the least number of websites were ownership of biological material and procedure for samples – disposal, which were covered by one apiece.

In total, 25 items were derived from professional recommendations. These included all four of the items covered by 12 websites, general privacy Issues, general privacy policy, confidentiality of test and data protection issues, and both items covered only by one website: ownership of biological

[±] This item is not included in the written description or statistics as it is not applicable to all of the websites.

Table 29 Coverage of Items in Privacy and Security Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
General privacy Issues	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	12
Privacy of data	✓		✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	11
Privacy of sample	✓				✓	✓	✓		✓	✓		✓	✓	8
General privacy policy	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	12
Privacy policy with regard to HIPAA[±]				✓	N/A			✓			✓	✓		4
Privacy with regard to health insurance	✓		✓		✓		✓		✓		✓	✓		7
Privacy with regard to the government	✓		✓		✓		✓		✓	✓	✓	✓	✓	9
Privacy with regard to potential employers	✓		✓				✓		✓		✓	✓		6
Privacy controls	✓		✓		✓		✓		✓	✓		✓		7
Confidentiality of test	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	12
Persons or groups with access to test results	✓		✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	11
Access to data or results with regard to third parties in general	✓		✓		✓		✓	✓	✓	✓	✓	✓	✓	10
Access to data or results with regard to insurance companies	✓		✓		✓		✓		✓		✓	✓		7
Statement that permission would be asked before third parties are given access to data or results[±]	✓		✓		N/A		✓		✓	✓	✓	✓		7
Instructions for accessing complaints procedure about breaches of privacy	✓											✓		2
Information about the sharing of data with outside parties	✓		✓		✓		✓	✓	✓	✓	✓	✓	✓	10
Statement about whether or not data can become part of medical records	✓		✓		✓						✓			4
(If information may be passed onto third parties) statement that information may be passed onto third parties	✓		✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	11
(If information may be passed onto third parties) statement of which parties information may be passed to	✓			✓	✓		✓	✓	✓	✓	✓	✓	✓	10
(If information may be passed onto third parties) statement with regard to the conditions under which information may be passed to third parties	✓		✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	11
Use of data by company	✓		✓		✓		✓	✓	✓	✓	✓	✓		9
Statement on the sale of consumer information or similar	✓		✓						✓		✓	✓		5

[±] These items are not included in the written description or statistics as they are not applicable to all of the websites.

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Information that is made public	✓		✓		✓					✓	✓		✓	6
Ease of identifying consumers from data	✓				✓									2
Information about whether or not data can be removed from company±	✓		✓		✓	N/A	✓		✓		✓	✓		7
Ownership of biological material							✓							1
Ownership of data	✓		✓				✓		✓	✓	✓			6
General fate of biological material	✓		✓	✓	✓	✓	✓		✓		✓	✓	✓	10
Fate of biological material if company sold or bankrupt±	✓		N/A			N/A	N/A		N/A					1
General security arrangements	✓		✓		✓		✓		✓		✓	✓		7
Security arrangements with regard to changes to administration	✓		✓				✓		✓			✓		5
General fate of data general	✓		✓		✓	✓	✓	✓	✓		✓	✓		9
Fate of data if company sold or bankrupt ±	✓					N/A	✓		✓	✓	✓	✓		6
Data protection issues	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	12
General security of data	✓		✓	✓	✓		✓	✓	✓	✓	✓	✓		10
General storage of data	✓		✓		✓		✓	✓	✓		✓	✓		8
Maximum period of sample storage						✓	✓		✓		✓	✓	✓	6
Maximum period of records storage					✓	✓	✓							3
Procedures for samples storage												✓	✓	2
Procedures for samples transfer														0
Procedures for samples disposal												✓		1
Procedure for records or data storage			✓		✓		✓		✓			✓		5
Procedure for records or data transfer			✓		✓		✓			✓		✓		5
Procedure for records or data disposal							✓		✓			✓		3
(If option of sending results by email) statement that sending results by email is not secure±	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A		N/A	N/A	N/A	N/A	0
Storage of information for statistical use	✓		✓					✓	✓	✓	✓	✓		7
Information about how results will be used	✓		✓		✓		✓	✓	✓	✓	✓	✓		9
Total	**36**	**1**	**32**	**9**	**30**	**11**	**35**	**18**	**34**	**23**	**32**	**38**	**17**	**316**

material and procedure for samples – disposal. Twenty-one items were derived from consumers' survey responses. These included two of the items covered by 12 websites, general privacy issues

± These items are not included in the written description or statistics as they are not applicable to all of the websites.

and general privacy policy, and one item covered by two websites: ease of identifying consumers from data.

The mean number of websites per item was 7.1. For those items derived from the recommendations of professional associations the mean was 6.9, and for those derived from consumers' survey results the mean was 7.7.

7.1.9 Tests and Testing Process

This section included all 29 items related to the tests, methods, processes or technology used (see Table 30).

The results showed that no website covered all 29 items. Lumigenix covered the most items, 26 in total. The only items not covered by Lumigenix were size of company's database or the number of users, quality control procedures and appropriateness of testing. However, no website covered size of company's database or the number of users, and only four covered quality control procedures. Appropriateness of testing was covered by eight websites however. Testcountry covered the least items, 10 in total. Seven of the items were covered by all 13 websites. One item, size of company's database or the number of users, was not covered by any website.

Eleven of the items were derived from professional recommendations, including five of the seven items that were covered by all of the websites and one of the items covered by only four websites: general technology and equipment used. Twenty items were derived from consumers' survey responses, including two of the items covered by all of the websites provision of analysis by the company and time taken and the item covered by no websites: size of company's database or the number of users.

The mean number of websites per item was 8.3. For those items derived from the recommendations of professional associations the mean was 11.2, and for those derived from consumers' survey results the mean was 6.7.

7.1.10 Counselling

This section included all 11 items related to counselling (see Table 31). Two of the items are not included in the written description or statistics as they were not applicable for all of the websites (see footnote underneath Table 31).

Table 30 Coverage of Items in Tests and Testing Process Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Provision of analysis by the company	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Nature of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Information about what the test entails	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓		12
General methods used	✓	✓	✓		✓	✓	✓		✓	✓	✓	✓		10
Genotyping methods used	✓				✓				✓	✓				4
Methods with regard to what data is compared to	✓		✓	✓	✓	✓	✓		✓		✓	✓		9
Methods of analysis	✓		✓						✓		✓			4
Probabilistic nature of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Characteristics of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Type of analysis	✓		✓		✓	✓	✓	✓	✓	✓	✓	✓		10
Information about how sample collected or how to do the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓		12
General technology and equipment used	✓				✓				✓	✓				4
Chip or platform used	✓		✓						✓	✓	✓			5
Information that is analysed	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓		12
Sample size for genetic associations	✓		✓						✓		✓			4
Size of company's database or the number of users														0
Depth or extent of analysis	✓		✓				✓	✓	✓		✓			6
Time taken	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Transparency of process	✓				✓				✓		✓			4
Algorithm for interpretation			✓						✓		✓			3
Quality control procedures	✓		✓					✓			✓			4
Method of sequencing	✓				✓				✓	✓				4
Limitations of sequencing method									✓					1
General purpose of testing	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Precise purpose of testing	✓	✓	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	12
Possibilities of testing	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Specific information about tests offered	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	12
Appropriateness of testing		✓	✓	✓	✓		✓			✓	✓	✓		8
Cost of test	✓	✓	✓	✓	✓	✓	✓	✓	✓		✓		✓	11
Total	**25**	**15**	**23**	**15**	**20**	**16**	**18**	**15**	**26**	**19**	**24**	**16**	**10**	**242**

The results showed that Navigenics covered all nine of the items, the only website to do so. Accumetrics/Viaguard, Geneplanet, International Biosciences and Lumigenix covered the least items, one apiece. Accumetrics/Viaguard only covered potential benefits from post-test counselling and the other three websites all only covered statement that tests may require interpretation by GP or genetic counsellor.

Table 31 Coverage of Items in Counselling Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Availability of counselling pre-test				✓		✓					✓	✓	✓	5
Availability of counselling post-test	✓		✓	✓		✓	✓			✓	✓	✓	✓	9
Potential benefits from pre-test counselling				✓		✓					✓		✓	4
Potential benefits from post-test counselling	✓	✓	✓	✓		✓	✓			✓	✓		✓	9
Cost of genetic counselling			✓			✓	✓				✓		✓	5
Recommendation for genetic counselling				✓		✓	✓				✓		✓	5
Contact details of genetic centres	✓			✓							✓			3
Statement that tests may require interpretation by GP or genetic counsellor			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	11
(If only available through healthcare professional or after counselling) should be made clear that only available through healthcare professional or after counselling[±]	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	✓	✓	N/A	2
Inclusion of counselling			✓	✓		✓	✓			✓	✓		✓	7
(If counselling is included) Information about whether consumers would be liable if they withdraw following pre-test counselling[±]	N/A	N/A	N/A	N/A	N/A		N/A	N/A	N/A	N/A		N/A	N/A	0
Total	**3**	**1**	**5**	**8**	**1**	**8**	**6**	**1**	**1**	**4**	**10**	**4**	**8**	**60**

No item was covered by all of the websites. The item covered by the most websites was tests may require interpretation by GP or genetic counsellor, which was covered by 11 websites. The item covered by the least websites was contact details of genetic centres, which was only covered by three websites.

All of the items in this section were derived solely from the recommendations of professional guidelines. The mean number of websites per item was 6.4.

[±] These items are not included in the written description or statistics as they are not applicable to all of the websites.

7.1.11 General Information

This section included all 18 items that were related to general information about genetics or were miscellaneous (see Table 32).

Table 32 Coverage of Items in General Information Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
Link to government health service websites with information about genetic testing										✓			✓	2
General information about genetics	✓	✓	✓		✓	✓		✓	✓	✓	✓	✓	✓	11
Role of genes in health and disease	✓		✓	✓	✓	✓		✓	✓	✓	✓	✓		10
Role of genes in conditioning phenotypes	✓		✓		✓	✓			✓	✓				6
Relative roles in determining health and disease of genetics	✓	✓	✓		✓	✓		✓	✓	✓	✓	✓	✓	11
Relative roles in determining health and disease of environmental factors	✓	✓	✓		✓	✓		✓	✓	✓	✓	✓		10
Relative roles in determining health and disease of lifestyle choices	✓	✓	✓		✓	✓		✓	✓	✓	✓	✓	✓	11
Relative roles in determining phenotype of genetics	✓		✓						✓	✓	✓			5
Relative roles in determining phenotype of environmental factors	✓		✓						✓		✓			4
Relative roles in determining phenotype of lifestyle choices	✓		✓						✓		✓			4
Appropriate information about useful health professionals	✓			✓			✓				✓	✓	✓	6
User reviews	✓		✓				✓	✓		✓	✓		✓	7
Bibliography	✓		✓		✓			✓			✓			5
Information about platforms that data can be accessed from			✓											1
Existence of an app														0
Difference between a completed DNA sequence and a list of SNPs	✓		✓						✓			✓		4
Information about what can be learnt from SNPs	✓		✓	✓	✓	✓		✓	✓	✓	✓	✓		10
Genetics of a specific disease	✓	✓	✓		✓		✓				✓			6
Total	**15**	**5**	**15**	**3**	**9**	**7**	**3**	**8**	**11**	**10**	**13**	**8**	**6**	**113**

The results showed that 23andme and deCODEme covered the joint most items, 15 in total. They both did not cover the items link to government health service websites with information about genetic testing and existence of an app. However, only two websites covered the first of these

items, and no website covered the second. 23andme also did not cover the item information about platforms that data can be accessed from, which was only covered by one website and deCODEme did not cover appropriate information about useful health professionals, which was covered by six websites. EasyDNA and Inherent Health covered the joint least items, three apiece. The only items covered by EasyDNA were role of genes in health and disease, appropriate information about useful health professionals and information about what can be learnt from SNPs. Inherent Health also covered appropriate information about useful health professionals, along with user reviews and genetics of a specific disease.

No item was covered by all of the websites. However, three of the items, general information about genetics, relative roles in determining health and disease of genetics and relative roles in determining health and disease of lifestyle factors, were covered by 11 of the websites. Only one item, existence of an app, was not covered by any website.

Eleven items were derived from the recommendations of professional associations. These included all three of the items covered by eleven websites, general information about genetics, relative roles in determining health and disease of genetics and relative roles in determining health and disease of lifestyle factors, and the item covered by two websites link to government health service websites with information about genetic testing. Seven items were derived from consumers' survey responses. These included one of the items covered by 10 websites, information about what can be learnt from SNPs, and the item covered by no websites: existence of an app.

The mean number of websites per item was 6.3. For those items derived from the recommendations of professional associations the mean was 7.3, and for those derived from consumers' survey results the mean was 4.7.

7.1.12 Coverage

This section included all 24 items that related to the coverage of the test (see Table 33).

The results showed that no website covered all of the items. 23andme covered the most items, 19 in total. Geneplanet covered the least items, seven in total. Six items were covered by all of the websites: what the test can say about health, what can be tested for or found out, scope of the test, type of test, general coverage of test and coverage of test with regard to conditions. Four items were not covered by any websites: comparison with regard to significance with SNPs not on chip, comparison with regard to significance with SNPs on other chips, density of coverage along the chromosome adjacent to a specific gene and number of SNPs tested within a specific gene.

Table 33 Coverage of Items in Coverage Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total
What the test can say about health	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
What the test can't say about health	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	12
What can be tested for or found out	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
What can't be tested for or found out	✓	✓	✓			✓	✓	✓	✓	✓	✓	✓	✓	11
Scope of the test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Genes tested	✓	✓	✓				✓				✓			5
Mutations or SNPs tested	✓		✓				✓	✓		✓	✓			6
Genes tested for a specific condition	✓	✓	✓				✓	✓			✓			6
Similarity of results between different companies											✓			1
Comparison with regard to significance with SNPs not on chip														0
Comparison with regard to significance with SNPs on other chips														0
Density of coverage along the chromosome adjacent to a specific gene														0
Amount of data that can be obtained	✓	✓	✓						✓		✓	✓		6
Number of SNPs tested	✓		✓						✓					3
Number of SNPs tested within a specific gene														0
Type of test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
General coverage of test	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Coverage of test with regard to conditions	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	13
Coverage of test with regard to number of conditions	✓	✓	✓	✓		✓			✓	✓			✓	8
Coverage of test with regard to carrier status	✓								✓			✓		3
Coverage of test with regard to amount of genome	✓								✓					2
Coverage of test with regard to parts of genome	✓		✓											2
Coverage of test with regard to comprehensiveness	✓		✓					✓	✓	✓				5
What the test can say about specific risk factors or susceptibility	✓	✓	✓	✓	✓	✓	✓	✓	✓		✓	✓		11
Total	**19**	**13**	**17**	**9**	**7**	**10**	**12**	**12**	**15**	**11**	**14**	**11**	**9**	**159**

Five of the items were derived from the recommendations of professional associations, including two of the items that were covered by all of the websites, what the test can say about health and

scope of the test, and one item covered by five websites: genes tested. Twenty-one of the items were derived from consumers' survey responses, including four of the items covered by all of the websites, what can be tested for or found out, type of test, general coverage of test and coverage of test with regard to conditions, and all four of the items not covered by any website: comparison with regard to significance with SNPs not on chip, comparison with regard to significance with SNPs on other chips, density of coverage along the chromosome adjacent to a specific gene and number of SNPs tested within a specific gene.

The mean number of websites per item was 6.6. For those items derived from the recommendations of professional associations the mean was 9.8, and for those derived from consumers' survey results the mean was 5.8 respectively.

7.1.13 Research

This section included all 22 items that related to the use of consumer data for research (see Table 34). Only companies that used consumers' genetic samples or data for research were included were assessed; the other companies are marked as not applicable. Since five companies were entirely not applicable, and other companies had several different items that they were not applicable for, percentage rather than absolute totals were used in the table.

All of the items were derived from professional recommendations. As the different companies were involved in different research activities, many of the items were not applicable for some of the websites, with a varied distribution throughout the different items and websites. Therefore the results could not be compared in the same way as for the other sections. Instead, percentages were calculated of the number of applicable websites that covered each item.

The results showed that Lumigenix covered the highest percentage of applicable items, a total of 56.3%. However, Lumigenix still did not cover seven applicable items. International Biosciences covered the lowest percentage of applicable items, with only 14.2% covered (18 not covered).

Aside from the item statement that samples or data are used for research (which was a prerequisite for inclusion in this section), no item was covered by 100 percent of websites. The item with the highest coverage was storage of data in a database with 87.5% (covered by seven websites). Two items, identity of third parties with access and transference of sample to a biobank, were not covered by any websites (out of a seven applicable websites each).

Table 34 Coverage of Items in Research Group

	23andme	Accumetrics /Viaguard	deCODEme	Easy DNA	Geneplanet	Genetic Health	Inherent Health	International Biosciences	Lumigenix	Mapmygene	Navigenics	Pathway Genomics	Testcountry	Total (%)
Statement that samples or data are used for research	✓	N/A	✓	✓	N/A	N/A	N/A	✓	✓	✓	✓	✓	N/A	**100.0**
Procedure for storage		N/A	✓		N/A	N/A	N/A		✓			✓	N/A	**37.5**
Procedure for disposal		N/A			N/A	N/A	N/A		✓			✓	N/A	**25.0**
Time period of storage		N/A			N/A	N/A	N/A				✓		N/A	**12.5**
Conditions of storage		N/A	✓		N/A	N/A	N/A		✓			✓	N/A	**37.5**
Identity of third parties with access		N/A	N/A		N/A	N/A	N/A						N/A	**0.0**
Information about possible lead to commercialization and patents	✓	N/A		✓	N/A	N/A	N/A						N/A	**25.0**
Customers' rights to commercial benefits	✓	N/A		✓	N/A	N/A	N/A	✓					N/A	**37.5**
Property of samples		N/A	N/A	✓	N/A	N/A	N/A		N/A				N/A	**16.7**
Property of data	✓	N/A	✓		N/A	N/A	N/A		✓	✓			N/A	**50.0**
Information about whether samples will be identified	✓	N/A	N/A		N/A	N/A	N/A		N/A	✓	✓	✓	N/A	**66.7**
(If samples are identified) how samples are identified	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	**N/A**
Destruction of samples	✓	N/A	N/A	✓	N/A	N/A	N/A		N/A		✓	✓	N/A	**66.7**
Communication of genetic risks		N/A			N/A	N/A	N/A			✓			N/A	**12.5**
Approval by ethics committee or similar		N/A			N/A	N/A	N/A		✓			✓	N/A	**25.0**
Transference of sample to a biobank		N/A	N/A		N/A	N/A	N/A		N/A				N/A	**0.0**
Storage of sample in a biobank		N/A			N/A	N/A	N/A		N/A		N/A	✓	N/A	**16.6666667**
Transference of data to a database		N/A	N/A		N/A	N/A	N/A			✓	✓		N/A	**28.57143**
Storage of data in a database	✓	N/A	✓		N/A	N/A	N/A	✓	✓	✓	✓	✓	N/A	**87.5**
Security measures	✓	N/A	✓		N/A	N/A	N/A		✓			✓	N/A	**50**
Potential risks	✓	N/A			N/A	N/A	N/A						N/A	**12.5**
Potential benefits	✓	N/A			N/A	N/A	N/A		✓	✓	✓	✓	N/A	**62.5**
Total (%)	**47.6**	**N/A**	**40.0**	**23.8**	**N/A**	**N/A**	**N/A**	**14.3**	**56.3**	**33.3**	**35.0**	**52.4**	**N/A**	

7.2 Analysis and Discussion of Content Analysis Results

7.2.1 Introduction

The analysis is split into three separate sections.

The first section compares the overall performance of the websites of the providers of DTC genetic tests assessed in the content analysis. This comparison is based on the number of items that each

website covered, and allows for an overall comparison across the range of information that companies should provide.

The second section compares the coverage of the different groups of items in the results. It also compares the coverage of the items that were derived from professional recommendations and those that were derived from consumers' survey responses.

The third section describes the coverage of the websites based on different themes, such as items to do with families or items to do with evidence, which cross the boundaries of the groups in the results. Important themes are described with reference to the coverage of items within them.

One issue that should be addressed at the start of the analysis is the lack of assessment of the quantity and quality of information provided about each item by the websites. Items were only assessed in a binary way; sufficient information about each was either provided or not (see Research Methodology). One obvious limitation of this method is that variations between websites in the information provided about individual items were not assessed. For example, two websites may have provided greatly differing quantities of information about a specific item, but if both were judged to have provided sufficient information then both would have been assessed as covering it. This limitation, however, was more than counteracted by the comprehensiveness, breadth and large number of the items assessed. In total, 284 items were assessed. This allowed for a large amount of detail to be examined. For example, the Accuracy group alone contained 28 items, covering a wide range of information from general information such as 'general accuracy of the test', to specific information such as 'sensitivity of the test'. Websites that provided only a small amount of information about accuracy would have been unable to cover a large number of different items, and correspondingly, websites which covered a large number of items would have needed to provide a reasonable quantity of information to be able to do so. Therefore, the total number of items covered by a website, or the number of items out of a particular group covered by a website, is a good indication of both the quantity of useful information and level of detail provided.

7.2.2 Comparison of DTC Company Websites' Overall Item Coverage

7.2.2.1 Total Coverage

The website that covered the highest percentage of items overall was 23andme, which covered 74.9% of items. This was closely followed by Navigenics, which covered 70.5% of items, and deCODEme, which covered 69.4%. As described in the previous section, these results show that these three websites had the best coverage of information on their websites, with regard to

professional association's recommendations and consumers' information need (see Methods for more information). In contrast, Accumetrics/Viaguard covered only 29.6% of the items, Testcountry covered only 29.8% and EasyDNA covered only 35.3%. These three websites therefore had the worst coverage of information. The coverage of the other seven websites was spread between these two groups.

One important point the results raise is the wide variation between the different companies: the website with the highest levels of coverage covered almost three quarters of the items and the website with the lowest covered less than a third. This variation is similar to that found in a previous content analysis of DTC genetic testing websites conducted by Lachance et al (2010), which found a similarly wide variation in both the quantity and quality of information provided by the different websites (a full examination of how the study results relate to previous research can be found in the Final Discussion). The results of this analysis support those findings, with many websites only covering the more general items in some areas, whilst others also covered more specific ones. It should also be noted that this wide variation invalidates a 'one size fits all' criticism of the information provided by the companies. For example, although some of the concerns raised in the literature (such as the overstatement of utility mentioned by Murray et al (2012)) are clearly important criticisms with regard to some of the websites, they may well be unfair in the case of others. This wide variation is also important when assessing the ethics of the tests. For example, a genetic testing service which clearly explains the important aspects of the test does not have some of the ethical problems, such as issues of informed consent, which a service which provides only limited information would have. Also, differences in the amount of information provided may affect consumers' opinions of the validity of the results, and could also affect any behavioural or psychological reactions to them.

A final point worthy of mention is that even the website which covered the most number of items, 23andme, still did not cover approximately a quarter of them. This finding suggests that a consumer who has purchased a test from the company which provides the highest level of information coverage on their website may still not have access to all of the areas of information suggested by professional recommendations and the survey respondents. Other websites covered substantially less material, which implies a lack of informed consent for, and understanding of, the tests for most consumers who rely on them as a sole source of information about the tests.

7.2.2.2 Rankings per Group

The previous section described the variation between the different websites' total coverage of the items in the content analysis. Although the total coverage is useful for an overall comparison of the websites, it does not illustrate the entire situation. When the items are split into their separate groups (as they are organised in the results) it can be seen that there was also variability in how well the websites covered the different groups. As different groups contained different numbers of items, the coverage of a small number of groups particularly well or badly may have influenced a website's performance with regard to total coverage, as described in the previous section.

Another way of comparing the websites is to examine how often the different websites covered a group particularly well, and how often they covered a group particularly badly. For every group of items (except the section on research) the websites were given a rank based on their coverage of the items in that group. The websites which were high-ranking (normally websites with the highest, second highest or third highest score) and the websites which were low-ranking (normally websites with the lowest, second lowest or third lowest score) were noted. Table 35 shows the number of times each website was high-ranking and the number of times each was low-ranking.

Table 35 Ranking of Website Information Provision

Website	High Rank	Low Rank
23andme	11	1
Navigenics	10	0
Lumigenix	9	1
deCODEme	7	0
Inherent Health	3	4
Pathway Genomics	3	4
Genetic Health	3	7
Geneplanet	2	4
Mapmygene	2	4
EasyDNA	1	7
Accumetrics/Viaguard	1	10
Testcountry	1	10
International Biosciences	0	6

The results show that several websites were consistently high-ranking. The two websites which were most often high-ranking were 23andme, which was high-ranking for all but one section, and Navigenics, which was high-ranking for all but two sections and was never low-ranking. This matches the order of the websites with regard to their total coverage of items (as described in the previous section), where 23andme and Navigenics were first and second respectively. However, deCODEme came third in the total coverage of items, but was fourth in the number of sections for

which it was high-ranking. Conversely, Lumigenix came third in the number of sections for which it was high-ranking, but fourth in the total coverage of items.

Similarly, several websites were consistently low-ranking. Accumetrics/Viaguard and Testcountry were the two most commonly low-ranking websites; both were low-ranking for all but one section. These two websites were also those which covered the lowest total number of items (as described in the previous section). The next two most commonly low-ranking websites were EasyDNA and Genetic Health, which were both low-ranking for seven sections. Although EasyDNA had the third worst coverage of the items in total (as described in the previous section), Genetic Health was only the fifth lowest. The fourth lowest was International Biosciences, which was the fifth most commonly low-ranking website in this section, as well as the only website to never be high-ranking.

It is interesting to note that nearly all of the websites had at least one section where they were high-ranking and one where they were low-ranking, with only three exceptions. Even the website which covered the highest number of total items and which was high-ranking for the most number of sections (23andme) was low-ranking for one section, and the two websites which covered the lowest number of total items and which were low-ranking for the most number of sections (Accumetrics/Viaguard and Testcountry) were high-ranking for one section. Other websites were in between, with high ranks for some of the sections but low ranks for others. Therefore, not only was there variation in the information provided between websites (as described in the previous section), but in most cases there was also variation in the information provided within websites.

7.2.3 Comparison of the Coverage of the Groups of Items

7.2.3.1 Benefits and Limitations

Although a full comparison of the results and the literature is given in the Final Discussion section, a useful study to mention here is that by Singleton et al (2012). Singleton et al performed a content analysis of 23 DTC genetic testing websites and found that the number of statements about the tests' positive aspects or benefits generally outweighed the number of statements about their negative aspects or limitations. This was quite an important finding as difference in coverage of benefits and limitations may affect consumers' opinions of the tests (as described below), and similar findings have been found in the current study. With regard to these issues, the main findings of the current research are as follows. The mean number of websites which covered each item in the benefits group was over double that of the limitations/risks group: 10.5 and 4.5 respectively. Although no website covered all 12 of the items in the benefits group, 23andme, Navigenics, Mapmygene and Pathway Genomics all covered 11 and deCODEme, Geneplanet, Inherent Health

and Lumigenix all covered 10. In contrast, all but three websites covered less than half of the items in the limitations/risks group. Each website covered a higher percentage of items in the benefits group than it did in the limitations/risks group. However, there was a wide variation in this difference. The website with the smallest difference was 23andme, which covered 84.6% of the benefits and 81.0% of the limitations/risks. In contrast Geneplanet, the website with the largest difference, covered 76.9% of the benefits but only 9.5% of the limitations or risks.

This difference between the coverage of the benefits and limitations/risks of the tests raises serious implications with regard to informed consent and consumers' understanding of the tests. In order for consumers to give consent for a test which is as fully informed as possible then they must be aware of all of its limitations and risks. For example, although all websites covered the 'limits to informative value' of the tests, and ten out of 13 covered general limitations of the tests, limitations of the tests with regard to claimed benefits and limitations to the interpretation of the results, only Pathway Genomics covered the consequences of investigation and only Navigenics the consequences of treatment. Seemingly important information, such as the risks of the tests with regard to discrimination for certain specific types of insurance (i.e. life, disability and long-term care) were only covered by two websites apiece, and even risks about insurance discrimination in general was only covered by five of the websites. As well as the doubts over informed consent that these findings raise, a lack of information on many of the potential limitations and risks of the tests, along with a correspondingly greater coverage of their benefits, risks giving consumers a false perception of the tests and should surely be considered against their general interests. However, it should be noted that 10 out of the 13 websites did provide information about the general limitations of the tests and all of them covered the limits to their informative value; consumers would therefore be not completely unaware of this topic.

7.2.3.2 Mean Websites per Item

Previous sections described a wide variation in item coverage between the different websites. However, as important as the differences between individual websites are, it is also important to analyse the coverage of the items by the websites as a whole. This allows for an assessment of which items are generally well covered and which poorly covered, and hence indicates which areas are important to the companies and on which areas consumers need more information. Although the level of coverage of each item is shown in the results, the large number of items precludes a comparison of each one individually. Therefore, a useful method is to compare the coverage of groups of items.

The items in the content analysis were organised into 13 appropriate groups, as shown in the Results section. In order to compare the coverage of the items in these groups, a score named 'mean websites per item' was created. The mean website per item is calculated as the sum of the number of websites that covered each item in a group divided by the number of items in that group. This gives a value for the mean coverage of items in each group, with a maximum of 13 (i.e. coverage of every item by each website). Table 36 shows the mean number of websites per item for 12 out of the 13 groups of items (the Research group was excluded due to the large number of items that were not applicable).

Table 36 Mean Websites per Item by Item Group

Group	Mean websites per item
Benefits	10.5
Tests and testing process	8.3
Results	7.9
Privacy and Security	7.1
Disease	7.1
Coverage	6.6
Counselling	6.4
General Information	6.3
Accuracy	6.2
Vendor	5.8
Limitations/Risks	4.5
Legal	2.3

As might be expected, the items in the benefits group were generally covered by more websites than any other area, with a mean websites per item score of 10.5, and the items in the limitations/risks group were amongst the least well covered, with a score of 4.5. As the primary purpose of the websites is to sell the tests, this is not surprising, and issues arising from this were discussed in the previous section. What is perhaps surprising is that the legal group had the lowest mean number of websites per item, and hence the worst coverage by the websites. Information about the legal aspects of the tests seems vital in order for consumers to be as fully informed as possible, and yet the only item covered by over half of the websites (10) was 'statement that third parties (e.g. law enforcement) may have access to samples if required by law'. Despite the concerns often raised with regard to insurance discrimination, the legal position of declaring results for (general) insurance purposes was only covered by five websites, with even less coverage for individual types of insurance. For example, no website covered the legal position of declaring results for medical insurance, and only three covered the legal position with regard to life insurance. Other important issues were similarly underrepresented. For example, only one website stated that taking

DNA from someone else (without consent) is a criminal offence in some jurisdictions, and no websites stated that this would be unethical.

After benefits, the two groups with the next highest scores for mean websites per item were tests and testing process and results, with scores of 8.3 and 7.9 respectively. Although these scores show the websites generally had a good coverage of the items in these groups, they are still substantially lower than the score for the benefits group. As with the benefits group, it is perhaps unsurprising that the websites tended to give the items in these areas a high coverage, as they describe the tests and results that consumers will purchase. For example, the items 'whether analysis would be provided by the company' and 'the nature of the tests' were covered by all of the websites, and all but one covered what the test entails and what to expect with the results: all important pieces of information about the product for sale. Their high coverage can also be explained by the fact that these groups focus more on positive aspects of the tests (i.e. what the company is providing) than negative aspects, with only a few exceptions.

The two groups with the next highest mean number of websites per item, with scores of 7.1 apiece, were privacy and security and disease. These are two important areas, and so it is welcome that they were amongst the groups with the most coverage. As above, the disease group is partly focused on the service provided by the company, helping to explain its relatively high level of coverage. For example, 'recommendations for appropriate actions based on results' were covered by all but two of the websites, and behavioural and environmental factors that will influence a condition were covered by nine each. Privacy and security is also partly about the service (i.e. security features provided) but also about the policy of the company towards consumers' information. Both of which are common concerns raised about DTC genetic tests.

The difference between the mean websites per item for the next five groups (coverage, counselling, general information, accuracy and vendor) was small, with a range of only 0.8. Apart from coverage, which was just over half, the items in these groups were generally covered by less than half of the websites. Although coverage and accuracy contained items which are thought to be very important for consumers to know, they also contained items that are highly technical and so may have been considered too complex for the general public. For example, all of the websites covered the scope of the test, what the test can say about health and how diagnostic they are. These relate to the service provided by the company, and so (as above) it is unsurprising that they are well covered. However, only three websites covered the sensitivity of the test and no website covered the specificity of the test or compared the significance of the SNPs tested with either SNPs not tested or SNPs tested on other chips. The lack of inclusion of these items is perhaps understandable, for the

reason given above, yet it is still disappointing with regard to consumers' understanding of the tests. Counselling is not provided by the majority of companies, which may explain why it is in the bottom half of the sections. However, it is an important area, and to give an example, only five websites covered the recommendation of genetic counselling, although nine did cover the potential benefits of post-test counselling. This low rank is possibly explained by the fact that the counselling group focuses less on what is provided by the companies than many of the other groups do.

7.2.3.3 Professional Recommendations and Survey Responses

As described in the Research Methodology, the items were derived from two different sources: the recommendations of professional bodies and the responses to the survey. Table 37 shows the mean number of websites per item for both sources in each group of items.

Table 37 Mean Websites per Item by Question Source

Group	Professional Recommendations	Survey Responses
Benefits	10.1	11.0
Tests and testing process	11.2	6.7
Results	8.3	7.6
Privacy and Security	6.7	7.7
Disease	7.0	7.5
Coverage	9.8	5.8
Counselling	6.4	N/A
General Information	7.3	4.7
Accuracy	6.6	5.7
Vendor	4.3	6.5
Limitations/Risks	4.6	4
Legal	2.0	6.0*

*only one applicable item in the section

For six of the groups the mean number of websites per item was higher for those items derived from the professional recommendations than for those derived from the survey responses. However, the items derived from the survey responses had a higher mean for five of the groups. This appears to show a balance in the websites' coverage of the information professional associations believe consumers should know and the information that consumers themselves wish to know. Delving deeper into the results shows some variability amongst the groups of items. Six of the groups have differences between the mean for the professional recommendations and the mean for the survey responses of 1.1 or under, displaying a good level of balance. However, the other five groups have much larger differences. One of these latter groups is the legal group. The difference for the legal group may be explained, though, by the fact that there was only one item derived from the survey responses for this group. The other four groups (tests and testing process, coverage, general

information and vendor), however, cannot be explained in this way. Interestingly, the vendor group is the only one of the four with a higher mean number of websites per item for the items derived from the survey responses. With regard to the other sections, it is possible that the companies' viewpoints are closer to the professional guidelines than to consumers, or they may have used the professional guidelines to decide what information to provide on their websites. Alternatively, it may simply be due to chance.

It is interesting to note that, with regard to the items in the coverage group, only one item derived from the professional guidelines mentioned SNPs compared to five items derived from the survey responses. The reason for this difference is unclear and can only be speculated upon. For example, since several of the items derived from the survey responses relate to information that would be useful when comparing different tests (e.g. 'comparison with regard to significance with SNPs on other chips') it is possible that consumers, who have to decide which test to purchase, may consider this information to be more important than professionals. Alternatively, as information about SNPs can be complex, professionals may not consider it to be appropriate information to provide for consumers. However, without further research no conclusions can be drawn and any difference may only be due to chance.

7.2.4 Coverage of Items Grouped Thematically

7.2.4.1 Analytical Features of the test

A large number of items from various groups dealt with the analytical features of the test. There is a wide variation in how well these items were covered.

There tended to be a higher coverage of the more general items. For example, the items relating to what the test can and cannot say about health, what can or cannot be tested for and the scope of the test were all covered by at least 11 websites. It is important to note that similar numbers of websites covered both what can be tested for or found out from the tests and what cannot. This equal coverage is perhaps surprising given the difference in coverage of the benefits and limitations/risks groups identified earlier, but is certainly welcome as it helps to create a balanced picture of the tests. The predictive value of the tests and how diagnostic the tests were (both coded as general rather than specific items) were both covered by all of the websites, as was 'general coverage of the test'. Two other general items, the probabilistic nature of the test and the highly nuanced nature of the results, were covered by 13 and 12 websites respectively. These are two particularly important items, as they deal with aspects of the tests that could be easily misunderstood. Adequate coverage of these items helps consumers to be aware that the results are

not absolute and that they need to be considered with regard to a range of other information. A similarly important item is 'statement of when results can only give relative, rather than absolute, risk'. However, this item had a lower coverage of just eight websites.

Not all of the more general items were quite so well covered as those described above. For example, the general scientific credibility of the results, general validity of the tests/science and the adequacy of the interpretation of the results were each not covered by three websites. The general accuracy of the tests was only covered by eight of the websites, as was the quality of, or confidence in, the analysis. It is interesting to note that the accuracy of the interpretations of the results or predictions was also only covered by eight websites, but the similar (and seemingly more negative) limitations of the interpretations of the results was covered by 10 websites; unusual given the expectation of a higher coverage of positive information. An even lower number of websites covered the amount of data that can be obtained (six), any variability in accuracy (five) and the accuracy of sequencing (four). The accuracy of sequencing and variability in accuracy are both items that it is important for consumers to know in order to fully understand the accuracy of the test; it is therefore concerning that so few websites covered them.

The items that dealt with more specific areas tended to be covered by fewer websites. This may be partly explained by the technical nature of some of the items. Also, for a website to cover a more general item it needed only to include information about something in that general area, rather than the more precise information needed for specific items. However, the lack of coverage of many of these items does add to the concerns about consumers' informed consent of and ability to fully understand the tests. For example, only two of the specific items that dealt with the analytical features of the tests were covered by more than half of the websites. These were the conditions that the test covers, which was covered by all 13 of the websites, and the number of conditions covered, which was explicitly stated on eight. These two items may have had high coverage because they dealt with the main features of the product. However, only three websites covered whether or not carrier status was included. The genes tested for a specific condition and the mutations or SNPs tested were each only covered by six websites, and the genes tested generally only by five. Although perhaps not important to all consumers, these three items are important for those who wish to delve deeper into what a company offers, and to compare it with other companies. Therefore the fact that under half of the websites covered them reduces the ability of consumers to do so, as does the lack of comparison on any website of the SNPs tested with SNPs not tested or SNPs on other chips, the coverage on only one website of the similarity of results between different companies and the low coverage of the comprehensiveness of the tests (covered by five), the number of SNPs

tested (covered by three), the amount of, or parts of, the genome tested (covered by two each), the number of SNPs tested within a specific gene (covered by zero) and the density of coverage along the chromosome adjacent to a specific gene (covered by zero).

Several of the specific items dealt with statistics on the accuracy of the tests. These were all poorly covered: the reliability or repeatability of the results was only covered by four websites, the error rate by three, the sensitivity (or proportion of correctly identified true positives) by three and the specificity (or proportion of correctly identified true negatives) by none. As above, this information may be considered too technical for many consumers. However, its lack seriously restricts consumers' ability to assess the differences between the tests. This also applies to a similar but non-statistical item, outside verification of coverage, which was only covered by three items.

7.2.4.2 Evidence

Six items from several groups related to evidence. These items were covered by a middle to low number of websites. The most covered item was evidence for interpretation of results, which was covered by eight websites. Also, evidence or links to evidence of the association between a genetic marker and a disease, condition or trait was covered by seven websites. However, only four websites referenced the criteria used to include or exclude literature. This is problematic, as not all published literature is of a suitable quality for the analyses conducted by the company. Therefore, if there is no way for consumers to know why evidence is included or excluded, it makes it difficult for them to assess the validity of the results. Similarly, only three websites covered 'scientific evidence available for population'.

Seven of the websites covered the fact that the results are based on current evidence which may change. Although it is welcome that seven websites did cover this, it is a serious problem for those websites that did not. This is one of the most important issues relating to DTC genetic tests, and concerns have been raised in the literature that consumers may change their behaviour, or be affected psychologically, based on results which themselves may change in the future as more evidence is discovered. The lack of coverage of this item by six of the websites raises serious doubts about the consumers of those websites having the ability to give consent to the tests that is as fully informed as possible.

Finally, only one website gave evidence for the benefits claimed by many of the companies. As the purported benefits of the tests were mentioned by all 13 websites, and are a major selling point, it is disappointing that this information was not provided. Although it should be noted that, as stated elsewhere in the thesis, there is currently little published evidence of these benefits.

7.2.4.3 Family

Eight of the items from various groups related to issues to do with families. These were all poorly covered.

The item which was covered by the greatest number of websites was 'statement that the results can be important for relatives', which was only covered by four websites. The risks of the tests to family members was only covered by two websites, and possible implications or consequences for family members was also only covered by two. No website stated that relatives have a right to know, that relatives have a right not to know or covered 'support for families'. Family-related issues are very important in DTC genetic testing as, since families share large amounts of their genetic code, risks for one family member may be very similar to those for another one. Similarly, any errors in the analysis for one family member will be passed on along with any results that are shared. As family members will not have given their informed consent to the tests, and will probably not have researched them, then this can be a serious problem. Also, it may be possible for an individual's genetic code to be identified from a family member's code. These issues are serious and sensitive, and it is unfortunate that they have not been properly covered.

The other issue relating to families is the testing of children. It is considered by many commentators that the unnecessary genetic testing of children is unethical, especially when the tests are not of a medical standard. However, no website stated that tests that do not meet clinical validity requirements should not be carried out in children, and only one website raised concerns over the testing of children (although one other website did mention that consumers had to be 18 or over). In fact, although it was not an item in the content analysis, more than one website explicitly allowed the testing of children, with one website actively encouraging it.

7.2.4.4 Methods of producing results

Sixteen items, mainly from the group entitled 'tests and testing process', dealt with the methods of analysing the samples and producing the results. This is perhaps not as important an area as some of the others: most companies would analyse the samples in a similar way, and it is unlikely that this information would have a major impact on consumers' decision as to whether or not to purchase the test. However, it is still useful information for consumers to properly understand the tests, and it is important for consumers to know the procedures for which they are giving consent.

There was a wide variation in the coverage of the items. The item that was covered by the most number of websites was 'information that is analysed', which was covered by 12. As this is a

fundamental piece of information about how the tests work it is unsurprising that it was highly covered. The type of analyses performed and the general methods used were both covered by ten websites, and what the data is compared to by nine. Again, the same explanation applies. However, the item with the next highest coverage was 'depth or extent of analysis', which was covered by six. This was followed by the method of genotyping, the method of analysis, the method of sequencing and quality control procedures, which were all only covered by four websites. These are areas which consumers need to know about if they are to properly understand the methods used by the companies, and it is therefore disappointing that so many websites did not cover them.

The chip or platform used and the general technology or equipment used were covered by five and four websites respectively. These are less important areas for consumers to know about for purposes of general understanding; although they may be useful for comparisons between the companies. The same is true for the sample size for genetic associations, algorithm for interpretation and the size of the company's database or number of users, covered by four, three and zero respectively. The limitations of the sequencing method were only covered by one website. It is notable that all but one website did not cover this, as, since it deals with the accuracy of the results it is arguably the most important item relating to the methods of producing results.

7.2.4.5 Presentation of and Access to Results

Various items, mainly but not exclusively from the results group, were related to the presentation of and access to the test results. These items were well covered by the websites, with only three covered by less than half. This high coverage may be due to the items' reference to the product sold by the companies; hence beneficial to advertise.

All of the websites covered the meaning of the results. This is unsurprising since, as described above, the results are the product which the company is selling, and the meaning is an important part of that. This is also true for 'is appropriate explanation of data provided', which was covered by all but one of the websites. The highly-nuanced nature of the results is an item that is of a high importance for consumers to know, and so it is to the companies' credit that it was covered by all but one website. What to expect with the results and the nature of data produced were both covered by large numbers of websites (11 and 10 respectively), again descriptions of the product sold by the company. The same is true for the manner in which test results will be provided, information provided and detail of results which were covered by nine apiece.

Two slightly more complex items had a middling level of coverage. These were 'statement of when results can only give relative, rather than absolute, risk' and what the results can be compared to;

both covered by eight websites. Relative risk is a ratio describing the difference in disease risk between two groups who differ in respect to a risk factor or factors (e.g. the presence of a particular allele) (Malenka et al 1993 and Baron 1997). In contrast, absolute risk describes the "absolute magnitude" of an individual or group's risk of disease (Malenka et al 1993, p.543). Awareness of this distinction is vital for understanding of the test results and it is therefore disappointing that five websites did not cover it.

Several of the items which had a lower level of coverage would probably not be as important as other items with regard to understanding the results, but are aspects of the tests in which consumers could be interested and possibly useful selling points for the companies. For example, the ability to match results with other users, the ability to share results and consumer access to raw data were covered by two, four and six websites respectively. Updates to interpretations, a description of the user interface and 'provision of updated test results to consumers' were covered by slightly more (seven), but still a surprisingly low number considering the potential benefits to the company of consumers having knowledge of this information. Equally surprisingly, a demo version of the results was only provided by eight websites. Given the fact that many of the major companies (such as 23andme) provided a demo version, it is surprising that many companies did not.

7.2.4.6 Privacy and Security Controls

A large number of items related to the privacy and security controls used by the companies.

All but one of the websites had a privacy policy. This is to be welcomed as the privacy and confidentiality of the results is a common concern raised about the tests. It is surprising that one website (Accumetrics/Viaguard) did not have a privacy policy, as it is such an integral part of the service. Of a similar level of importance are data protection issues, which were again covered by all but Accumetrics/Viaguard.

Ten of the websites covered the general security of data, eight covered the general storage of data and seven covered the general privacy controls. These are reasonable numbers, although it is disappointing that several websites did not cover these general items. Also, the coverage was much lower for specific procedures: only five websites covered the procedure for records or data storage, five covered their procedure for transfer and only three covered their procedure for disposal. The coverage of sample procedures was even lower, with only two websites covering the procedure for their storage, one for their disposal, and zero for their transfer. These are important pieces of information for consumers to know if they are to understand the privacy and security controls as fully as possible, and their lack of coverage indicates that most consumers who rely solely on DTC

company websites for information would not be as fully informed as possible in this area. Similarly, only six websites covered the maximum period of sample storage, and only three the maximum period of records storage.

Seven websites covered whether or not data could be removed from the company. Seven websites also covered the general security arrangements; not large numbers but over half of the websites. However, only five websites covered security arrangements with regard to changes in administration. This is important information, as consumers' personal information needs to be secure for their lifetime (and possibly that of their families), not just the lifetime of the company.

7.2.4.7 Interventions

Several items from various groups related to interventions to improve health behaviour.

Perhaps unsurprisingly, items relating to negative aspects of interventions had poor coverage. For example, consequences of investigation and consequences of treatment were both only covered by one website. Although unsurprising, it is still disappointing as this is important information for consumers to know if they are fully to understand any behavioural changes they plan to make. Excluding this information must be considered unethical, as consumers would only be informed of the benefits of an intervention, not the consequences. Similarly, evidence for recommended interventions was only covered by one website and evidence against by none.

The positive items had much higher coverage. Whether results can help inform health behaviour choices and how they can do so were both covered by all of the websites. As in other sections this is unsurprising, as these areas are amongst the main selling points of the tests. Similarly, recommendations for appropriate actions based on results, specific applications of the results, behaviour that influences development of a condition, which results are actionable and possible prevention strategies were all reasonably well covered (by 11, 10, nine, eight and seven respectively).

One item that had notably good coverage was that results should not be used alone for medical decision-making. This is vital information for consumers to know, and it is welcome that it is covered by all but one of the websites.

7.2.4.8 Ethnicity

Three items related specifically to ethnicity or population. Although this is a small number of items, it is a particularly important area. Most associations in DTC genetic tests are based on research

involving European populations, which means that many associations may not be valid (or are unconfirmed as valid) for people with non-European ancestry.

Only four websites covered the populations for which the information about gene-disease associations is known; three covered the scientific evidence available for these populations and four covered the populations at risk of specific diseases. This is a serious lack of information from the majority of websites, which makes it difficult, if not impossible, for consumers from non-Caucasian ethnicity to determine how applicable the results are for them.

7.2.5 Summary

The content analysis was a comprehensive study with a large and detailed collection of items, which has produced several important findings. Firstly, a large variation was found between the coverage of the items by the different websites, and hence, of the information provided on them. Secondly, and notably, even those websites with the best information coverage still did not cover a substantial portion of the items, and those with the lowest level of coverage did not cover a large majority. Thirdly, there was a large variation in the coverage of different groups of items within the websites; although some were high-ranking for most groups and some were low-ranking for most groups, nearly all of the websites had at least one group where they were high-ranking and one where they were low-ranking. Fourthly, despite variation in some of the groups of items, the overall coverage of items derived from professional recommendations and those derived from the survey responses was broadly balanced. Finally, although many important items were well covered by the websites, a substantial number were subject to poor coverage.

Overall, the results raise serious issues about the ability of consumers fully to understand DTC genetic tests and to give consent that is as fully as possible informed based on the information provided on the websites of providers.

8 Final Discussion

8.1 Introduction

The aim of the research was to investigate the informational aspects of direct-to-consumer genetic tests, including the provision of information by the test providers, consumers' information needs and information-seeking behaviour and the effect of the information generated by the tests on health behaviour and health anxiety. This aim was designed to fill the research need identified after a review of the literature, and required the formulation of six research questions and 14 research objectives (see Chapter 1).

In order to answer the research questions three studies were conducted: a survey, email interviews and a content analysis. The results of each study have been analysed and discussed individually in Chapters 5, 6 and 7. However, although each study can stand alone on its merits, the three studies were interlinked (see section 4.1.2.3); therefore this chapter brings together the most important findings and discusses them as a whole, with a focus on how individual findings relate to each other, compare with the literature and answer the research questions.

8.2 Health Behaviour and Health Anxiety

The first two research questions were focused on health behaviour and health anxiety:

1. What effect does the information from a DTC genetic test have on consumers' health behaviour and health anxiety?
2. How does the information from a DTC genetic test effect consumers' health behaviour and health anxiety?

These questions were examined by the completion of objectives 1-4 (see Chapter 1) in the survey and email interviews:

1. To identify a sample of consumers of DTC genetic tests and a sample of individuals who are interested in the tests but have not yet purchased one.
2. To inquire into changes to consumers' health behaviour and health anxiety after receipt of DTC genetic test result information.
3. To compare the current health behaviour and health anxiety of consumers of DTC genetic tests with individuals who are interested in purchasing a test or who have purchased one but not yet received their results.

4. To assess the mechanisms through which the information from a DTC genetic test can affect health behaviour and health anxiety.

8.2.1 Health Behaviour

Several findings are useful in answering the health-behavioural aspects of the research questions. Firstly, no participant in the research reported any cessation of an existing health behaviour, even amongst those whose results had shown a decreased risk of disease. This was one of the concerns highlighted in the literature, and so it is important that no evidence has been found for it. Although this finding cannot be considered conclusive, it is consistent with previous research in the literature. For example, although Bloss et al (2010, 2011b, 2013) did not analyse participants' health behaviour individually, they did find that, as a whole, there was no significant decrease in participants' exercise behaviour or significant increase in their fat intake after exposure to the information in their test results. Kaufmann et al (2012) stated that none of their respondents reported a decrease in exercise behaviour; although they did not explicitly rule out any decrease in, or cessation of, any other health behaviour, none was mentioned in their results. Similarly, no cessation or decrease was mentioned by Gordon et al (2012) or Wasson et al (2013) when describing health behavioural changes amongst their participants. This agreement with the literature adds weight to the findings, and although, as stated above, they cannot be considered conclusive, the evidence does point towards a lack of negative effects of the information generated by the tests on consumers' health behaviour.

Secondly, both methods used to investigate the effects of the information from the tests on health behaviour (see section 4.3.2.1.2) found a broadly positive effect: when asked directly, a sizeable minority of consumers stated that their health behaviour had positively changed due to receiving their results; when the overall adherence of consumers and potential consumers to six common health behaviours was compared, it was found that consumers had a significantly higher level. The combined use of these two methods strengthened the findings, since their strengths and weaknesses were complementary (see section 4.3.2.1.2). Results were broadly in agreement with those of Kaufman et al (2012) and Gordon et al (2012), who found that a large minority of participants reported positive changes to their health behaviour. However, it differs from the study by Bloss et al (2011b), who found no significant changes in exercise behaviour or fat intake. This discrepancy may be due to a difference in methodology: this research assessed six common health behaviours, and, similarly to Kaufman et al (2012) and Gordon et al (2012), used open-ended questions. In contrast, Bloss et al (2011b) used no open-ended questions and only assessed two health behaviours. Indeed, this possibility is strengthened by the diverse range of different positive changes to health behaviour reported in the survey, many of which would not have been included in

Bloss et al's assessment. Due to the lack of experimental manipulation or pre-test health behaviour assessments these findings cannot prove causation. However, when combined with the results of the studies by Kaufman et al (2012) and Gordon et al (2012), they give a strong indication that the tests have a positive effect on the health behaviour of a minority of consumers. There are some limitations of the research that should be noted when considering these findings (see section 9.3). However, the strengths of the research complement the limitations of the previous studies, and vice versa, allowing for a fuller picture of the area. For example, it was necessary to use a convenience-sampling method (see section 4.3.2.2), which may have introduced bias. However, this allowed for the inclusion of 'real' consumers, a key missing element of the studies by Bloss et al (2011b) and Gordon et al (2012) (see Methods for other examples).

Thirdly, respondents generally reported a reasonable approach to the information in their results. Many of those who reported behavioural changes made specific changes for specific high risk areas. For others, the change was a mixture of the information in their results and other factors, such as family history. Similar to the findings of Wasson et al (2013), the information generated by the tests was often seen as giving a motivational push, with several respondents adopting health behaviours that they had already known were a good idea. Many of those who did not change their behaviour reported that there were no areas of concern in their results, but others were sceptical about their usefulness or the seriousness of reported disease risks. There was no evidence for extreme reactions to the information generated by the tests, more a general impression that they were a useful tool. These findings are consistent with those of Gordon et al (2012), who found that most of their participants had a reasonable understanding of, and approach to, their results.

8.2.2 Health Anxiety

The findings about health anxiety are much less clear-cut than for health behaviour. The majority of respondents stated that their health anxiety did not change due to receiving the information in their results. However, some respondents did report a change: most a decrease but a small number an increase. Despite these reported changes, there was no significant difference between consumers' and potential consumers' current health anxiety levels; possibly due to the small magnitude of most of the reported changes to anxiety.

Any increase in anxiety is a cause for concern, especially given the "psychosocial distress" observed by Mahon (2012, p.260) in three patients whose results showed a high risk of cancer. However, it is reassuring that 138 of the 183 consumers who answered the survey questions on health anxiety reported no change to health anxiety. Also, out of the 34 consumers who described how their

health anxiety had changed only five reported an increase; two of whom also reported a decrease. The three respondents whose anxiety solely increased unfortunately could not be included in the interviews; they either did not give an email address or did not wish to participate. The magnitude of these increases could, therefore, not be fully analysed. However, two of the three did mention the magnitude in their survey responses, and indicated that the increase was small.

In terms of health anxiety, as in health behaviour, respondents generally reported a reasonable approach to the information in their results. A decrease in anxiety was often described as due to a lack of an increased risk for a disease that ran in the family, an increase in knowledge about their health as a result of the information provided by the tests and/or a sense of empowerment caused by the information in their results. One respondent did report that his anxiety decreased because he had low risks for diseases that ran in his family; a possible cause for concern due to the uncertain accuracy of the results i.e. the respondent might actually have a high risk. However, this individual stated explicitly that his results did not preclude him from contracting the disease. As stated above, the majority of respondents reported no change in health anxiety. The main reasons reported for this lack of change were to do with respondents' attitudes towards health and the tests. These showed a generally level-headed approach, an awareness of the limitations of the tests and an understanding of how genetic risk estimates correspond with general risks in life.

The reported changes to health anxiety differ from Bloss et al's (2011b, 2013) study, which found no significant difference in the pre- and post- test health anxiety levels of their 2037 participants. It is likely that this is due to the difference in methodology, as Bloss et al analysed participants as a whole, rather than individually. Indeed, the lack of significant difference between the health-anxiety scores of consumers and potential consumers in this research backs up this idea; changes to anxiety may be too small to measure other than individually. However, the results are similar to Gordon et al's (2012), who found that most participants had a moderate response, with no large increases in anxiety. Interestingly, 25% of the 60 participants in their study stated that they were worried about their results; a much higher proportion than the five respondents in this research who reported an increase in health anxiety. This difference may be due to a difference in the language used i.e. a change in anxiety may be considered to be more serious than simply being worried. Alternatively, it is also possible that it is due to the lack of 'real users' in Gordon et al's study (as described previously). It is also interesting to compare the five respondents who reported an increase in anxiety in this research with the fact that 40% of the 319 participants in the study by Bansback et al (2012) predicted that their anxiety would increase after receipt of test results. However, since those participants were only provided with hypothetical test results a difference is unsurprising.

The findings of this research and of previous studies suggest that concerns about large increases in health anxiety are unfounded, and that most consumers will either have no change to their health anxiety due to the information provided by the tests or will have a reduction. However, research so far is far from conclusive, and more is needed before firm conclusions can be drawn.

8.3 Information

The third to fifth research questions were focused on information about the tests:

3. What are consumers' information needs and information-seeking behaviours?
4. What effect does the information from a DTC genetic test have on consumers' information needs and information-seeking behaviours?
5. Are consumers' information needs met by the information provided on the websites of companies that sell DTC genetic tests?

These questions were examined by the completion of objectives 1 and 5 to 11 in the survey, email interviews and content analysis:

1. To identify a sample of consumers of DTC genetic tests and a sample of individuals who are interested in the tests but have not yet purchased one.
5. To assess the information needs of consumers of DTC genetic tests.
6. To assess the information-seeking behaviours of consumers of DTC genetic tests.
7. To assess changes to consumers' information needs after receipt of DTC genetic test result information.
8. To assess changes to consumers' information-seeking behaviours after receipt of DTC genetic test result information.
9. To identify all providers of DTC genetic tests for multiple disease risk assessment.
10. To identify recommendations for the information that should be provided by providers of DTC genetic tests.
11. To analyse the information provided on the websites of providers of DTC genetic tests with regard to recommendations of information that should be provided and consumers' self-identified information need.

One of the main contributions of this research is the identification of consumers' information needs from their point of view. As described previously, this is a large gap in the literature; only consumers' information needs from the professional viewpoint have been described. As might be expected from a large and diverse group of individuals, reported information needs were highly

varied. The most commonly-cited needs were coverage and accuracy, but cost, privacy and information about the results and samples were also very important, along with a wide range of other topics.

Respondents themselves reported a reasonable level of contentment with the information provided on the websites of providers of DTC genetic tests. For example, when asked to rate from one to seven (low to high) how much they agreed with the statement 'I am generally satisfied with the information provided on the website', the mean rating was 5.75. Contrary to this, the content analysis showed that information provision on the websites was generally poor; a finding strengthened by its agreement with the previous content analyses in the literature by Lachance et al (2010), Hennen et al (2010), Geransar and Einsiedel (2008) and Liu and Pearson (2008).

The results from the content analysis were another important contribution. Although there have been several other content analyses of providers' websites (see section 2.4.2.2), this has been the only analysis to have included items based on information that consumers wish to know. As stated above, the content analysis found that the information provision on providers' websites was generally poor. However, there was a wide variability in the quality of information provision, as found by Lachance et al (2010). The website with the highest coverage of items covered 74.9%, vastly different to the 29.6% covered by the website with the lowest coverage. This finding implies that choice of provider greatly influences the information that consumers will receive; although whichever provider is chosen will still not provide information on a fair number of topics. The coverage of items also varied greatly depending on the topic. Unsurprisingly, the topic with the highest mean coverage was the benefits of the tests, which had over double the coverage of the limitations of the tests. This finding is similar to the results of a study by Singleton et al (2012), who found that, on average, websites had six times as many benefits statements as statements of risks and limitations. It is therefore reasonable to conclude that the information that consumers receive from providers' websites is generally skewed to give an inaccurate picture. It is also concerning that the topic with the lowest coverage was found to be legal issues, and that other areas important to the understanding of the results, such as the effect of ethnicity on disease risk, were poorly covered.

It is important to note, however, that the information provided by the websites is only one side of the story. The interviews showed that consumers do not just rely on providers' websites for their information, but also seek it from other sources. Consumers' information-seeking behaviour varied widely; some extensively sought information whilst others spent only a short time seeking it. Although many of the sources used could not be considered suitably scientific, it is reassuring that, based on the interview sample, most consumers searched for information about the tests before

purchase. These findings do relieve some of the concerns about the poor information provision of the providers, as most consumers will not rely solely on it. They also call into question the examination of these issues in the literature; although many studies have analysed the information provision of the websites, and many commentators have raised concerns over consumers' informed consent, the information-seeking behaviour of consumers has not previously been studied. This dislocation between researchers and consumers is further emphasized by the finding that the majority of interview respondents felt that consumers and providers share the responsibility for ensuring that consumers are fully informed, rather than the providers alone. However, despite these findings, concerns in the literature are partly justified; most consumers, based on the interview sample, only search for information about a small range of topics. Therefore, it is indeed likely that, due to the poor information provision of the providers, most consumers are not as fully informed as possible about topics that they do not search for themselves.

The findings about the effect of DTC genetic tests on information need and information-seeking behaviour are much less clear. Although interview respondents stated that they kept up with the area, few reported extra information that they had searched for after receiving their results. Some consumers in the survey did report further information needs after receipt of results, but this was only a small minority.

8.4 Other

Aside from the concerns in the literature mentioned throughout the thesis, this research was an ideal opportunity to examine consumers' opinions about the tests. Therefore the final research question was:

1. What are consumers' opinions about, and experiences with, DTC genetic tests?

This question was examined by the completion of objectives 12 and 13 in the email interviews:

12. To assess consumers' opinions about DTC genetic tests.
13. To assess consumers' experiences with DTC genetic tests.

Consumers had a wide variety of different opinions and experiences.

For example, respondents' reasons for purchasing a test included genealogy, health, curiosity and novelty. This is similar to the results of a study by Su et al (2011, p.135), who found that respondents' main motivations and expectations were "health...curiosity and fascination...genealogy....contributing to research and....recreation".

Respondents varied in their opinion about the genetic knowledge an individual should have before purchasing a test; some considered a basic level important, others that it was unimportant, dependent on provider used or that knowledge of statistics was more important. Similarly, respondents varied in attitudes towards regulation, some were in favour and others strongly opposed; findings which are similar to Bollinger et al (2013), who found that two thirds of consumers were against regulation but most favoured oversight of the scientific validity of providers' claims.

Previous studies have generally found a majority of respondents predicting that they would share their results with a healthcare professional; 78% in McGuire et al (2009), 92% in Gollust et al (2012) and 63% in Bansback et al (2012). This is consistent with the interview findings, where 17 respondents stated that they had shared results and 10 that they had not.

These are but a few examples of opinions and experiences identified in the research, mainly in the email interviews. They are an important contribution as they bring the views of consumers into a debate from which they have not been completely excluded (as with information need), but have been underrepresented.

8.5 Ethical Implications of Research

8.5.1 Introduction

A comprehensive analysis of the ethics of the provision of DTC genetic tests is beyond the scope of this thesis. However, to the author's knowledge the literature is without an ethical analysis of the tests based on actual findings, and the breadth of this research provides a good opportunity to briefly examine the ethical implications of the findings with regard to the provision of DTC genetic tests.

As described in section 2.4.1.3, the legal position of DTC genetic tests varies from country to country, and has generally yet to be conclusively settled. For example, the FDA in the USA is currently considering whether or not to grant 23andme approval for a selection of its tests. An analysis of the adoption of a screening test by the medical profession would traditionally focus on the pros and cons of its adoption, with minimum thresholds for benefits and maximum thresholds for risk. However, it is important for this analysis to be grounded in reality. The current situation is that, in most countries, DTC genetic tests are legally available. Therefore the matter is not as simple as a question of the ethics of their provision. If a jurisdiction in which they are currently permitted considered their provision to be unethical and decided to introduce a ban, it would be an active choice; rather than simply preventing the tests from becoming available, they would be removing a

service that the public has access to; a decision that may in itself cause harm. Therefore, the ethical implications need to be examined from two viewpoints: the ethics of the provision of the tests and the ethics of a ban on the provision of the tests.

The ethical issues are examined based on utilitarianism, respect for autonomy and nonmaleficence (Beauchamp & Childress 1994, pp.47-48, 120-121, 189-192).

8.5.2 Utilitarianism

Utilitarianism is based on the principle that the ethical action is "the one that produces the best overall result" in terms of "good and bad consequences" (Beauchamp & Childress 1994, p.47).

As described in earlier sections of this thesis, there are concerns that the provision of DTC genetic tests may cause harm to consumers. However, both this research and previous studies have found no evidence of negative effects of the tests on health behaviour. On the contrary, positive effects have been observed in a minority of consumers. In terms of health anxiety, a negative effect has been observed in a very small proportion of consumers, but a positive effect in a much larger minority. It seems clear therefore that, for these two issues, current research indicates that the provision of the tests causes more good than harm. However, with regard to the provision of information about the tests the situation is less clear. The research indicated that, similar to the findings of previous studies, the information provision on providers' websites is poor. However, what the research did not assess was whether this poor information provision causes harm to consumers. It is certainly possible that it does so, but it is also possible that the information that is provided may benefit consumers, causing curiosity about health and a desire to seek further information. Therefore, this particular issue requires further research to clarify.

If a ban on DTC genetic tests was introduced, individuals would be prevented from experiencing the positive effects of the tests on health behaviour and anxiety that this research indicates. This would be a clear negative consequence of such an action. Although a ban would have some positive effects, namely in the prevention of an increase in health anxiety in a small minority of consumers, and the removal of the possibility of consumers ceasing any health behaviour due to good results, these would be far outweighed by the negatives. Once again, the situation with regard to information provision is uncertain; a ban would remove the possibility of poor information provision causing consumers harm, but remove the possibility of the information creating curiosity about health and a desire to seek further information.

8.5.3 Nonmaleficence

Nonmaleficence can be considered as an obligation "not to harm others" (Beauchamp & Childress 1994, p.190).

Research so far has found no evidence that the provision of DTC genetic tests violates the principle of nonmaleficence in terms of health behaviour; as stated numerous times, there is no evidence that the test results have a negative effect on health behaviour. However, it should be noted that this has not been proven, and it is possible that further research may identify harmful effects. Also, as described throughout the thesis, there are still many aspects of DTC genetics tests that have the potential to cause harm, such as the inherent uncertainty over the accuracy of health reports based on only part-sequencing of the genome and as yet incomplete genetic knowledge. Therefore, despite the lack of evidence for violation of this principle with regard to health behaviour, it cannot be proven that no violation occurs. With regard to health anxiety, a small amount of harm has been identified. When combined with other research in the literature, such as the psychological harm reported by Mahon (2012), it is possible that the tests' effects on anxiety are unethical in terms of nonmaleficence. However, there is not yet sufficient evidence to prove this and further research is required. The effects of the poor information provision identified in this research are, as described above, currently unknown. However, it is perfectly possible that some harm may be caused by it and that providers' lack of satisfactory information provision may violate the principle of nonmaleficence.

Although the provision of DTC genetic tests may cause some violation of the principle of nonmaleficence, it is considered that a ban on provision would likely create a much greater violation. As stated previously, a ban would remove the possibility of individuals benefitting from the observed positive effects of the tests on health behaviour and health anxiety; clearly a harmful action. In an extreme case reported in this research, one consumer may have died if her test results had not encouraged her to ask for a cancer biopsy; if a ban had been in existence before her purchase she therefore may not have survived. Once again, the effect of a ban on the test with regard to information provision is unknown, but the possibility of harm certainly exists.

8.5.4 Respect for Autonomy

The principle of respect for autonomy is based on the idea of respecting an individual's choices about their health and wellbeing (Beauchamp & Childress 1994, p.120).

The ethical situation appears mixed with regard to respect for autonomy. The provision of DTC genetic tests clearly allows people to make their own choices about whether to purchase a test or not. This allows them to discover information about their health which may otherwise be unavailable. Several respondents in the interviews described how this information allowed them to take control of their health, thus increasing their autonomy. However, the content analysis, along with similar studies in the literature, has identified the information provision on providers' websites to be poor; even the best websites still did not provide information about a large number of issues. Although many respondents searched for information themselves, this was focused on a small number of issues rather than a comprehensive enquiry about the tests. Therefore, it is likely that many consumers are not as fully informed as possible when purchasing a test; this makes it difficult for them to exercise full autonomy.

With regard to a ban on the tests, the issues are obviously reversed. The lack of poor information provision does not have the effect of calling consumers' autonomy into question. However, a ban on a test which many individuals wish to take clearly reduces their autonomy, which is unethical in terms of this principle.

8.5.5 Summary of Ethical Implications

Further research and a more detailed ethical examination of the tests are clearly needed. Assuming future research does not identify a large negative effect of the tests on consumers' health behaviour or health anxiety, and that it is not discovered that the poor information provision causes harm to consumers, it seems likely that provision of the tests is ethical in terms of utilitarianism. In regard to nonmaleficence and respect for autonomy the ethical situation is currently unclear, although the evidence points towards possible violations with regard to the former.

However, any ban on DTC genetic tests would be an active decision, removing a service that is currently available to consumers in most countries. This brief examination of the ethical issues seems clear that, unless further research reveals surprising results, the act of banning the tests would be unethical in terms of utilitarianism, nonmaleficence and respect for autonomy.

8.6 Two Models of DTC Genetic Tests

The final objective in this research was:

14. To develop a model or models to describe the research findings with regard to consumers' experiences when purchasing a DTC genetic test and their related information-seeking behaviour.

After reviewing the findings it was considered appropriate to develop two models. The first model is one of the purchase of a DTC genetic test and the second of the information-seeking behaviour of consumers of DTC genetic tests.

8.6.1 Model of the Purchase of a DTC Genetic Test

As described above, the research findings allowed for the development of a model of the purchase of a DTC genetic test. As parts of the research were highly exploratory (such as the identification of consumers' information needs and information-seeking behaviour) the model is not designed to be comprehensive. Neither should it be considered a blueprint, wherein each individual adheres to every component and connection. Rather, it highlights the different processes, interactions and results of the purchase of a DTC genetic test found in this research. It is a simplified model, with many components combined, in order to present this in a clear and understandable way. Figure 14 shows this model.

The process of purchasing a test is shown as composed of five stages: discovery of the tests, interest in purchasing a test, intention to purchase a test, purchase of a test and receipt of results. Although some of these stages are similar, the results justified this distinction due to the specific events and interactions experienced by some consumers at the different stages. For example, most respondents who changed their behaviour did so after receipt of results. However, for one individual the act of purchasing a test itself was a motivational boost.

The 'process of purchasing a DTC genetic test' component is linked to the 'information needs, information seeking' component. Any stage of the process of purchase can generate information needs, which in turn can influence information seeking. The model shows the information needs and information sources most commonly cited in the survey. However, these are just a small selection of the large variety of information needs and sources described by respondents; each consumer and potential consumer will have their own individual set of needs and search methods, influenced by their existing knowledge, opinions and attitudes about various subjects. The results of the information seeking may influence the process of purchase. The extent to which this occurs is highly dependent on the individual; the interviews illustrated that some respondents were convinced to purchase a test by the information that they had found, whereas others had decided to purchase a test before seeking information. The information-seeking process may also stimulate more information needs, which would in turn stimulate further information seeking.

The model shows that each stage in the process of purchasing a test can be influenced by the individual's knowledge, opinions and attitudes about various topics. This is a separate component to

information needs and information seeking; it is composed not just of the information that an individual has discovered in the information-seeking process, but also their prior knowledge, opinions and attitudes. This is an important distinction; the research demonstrated that existing knowledge and opinions can influence individuals' opinions about the tests as well as affecting the way in which the information that they find influences their interest in and decision to purchase. Existing knowledge, opinions and attitudes can also influence individuals' information needs.

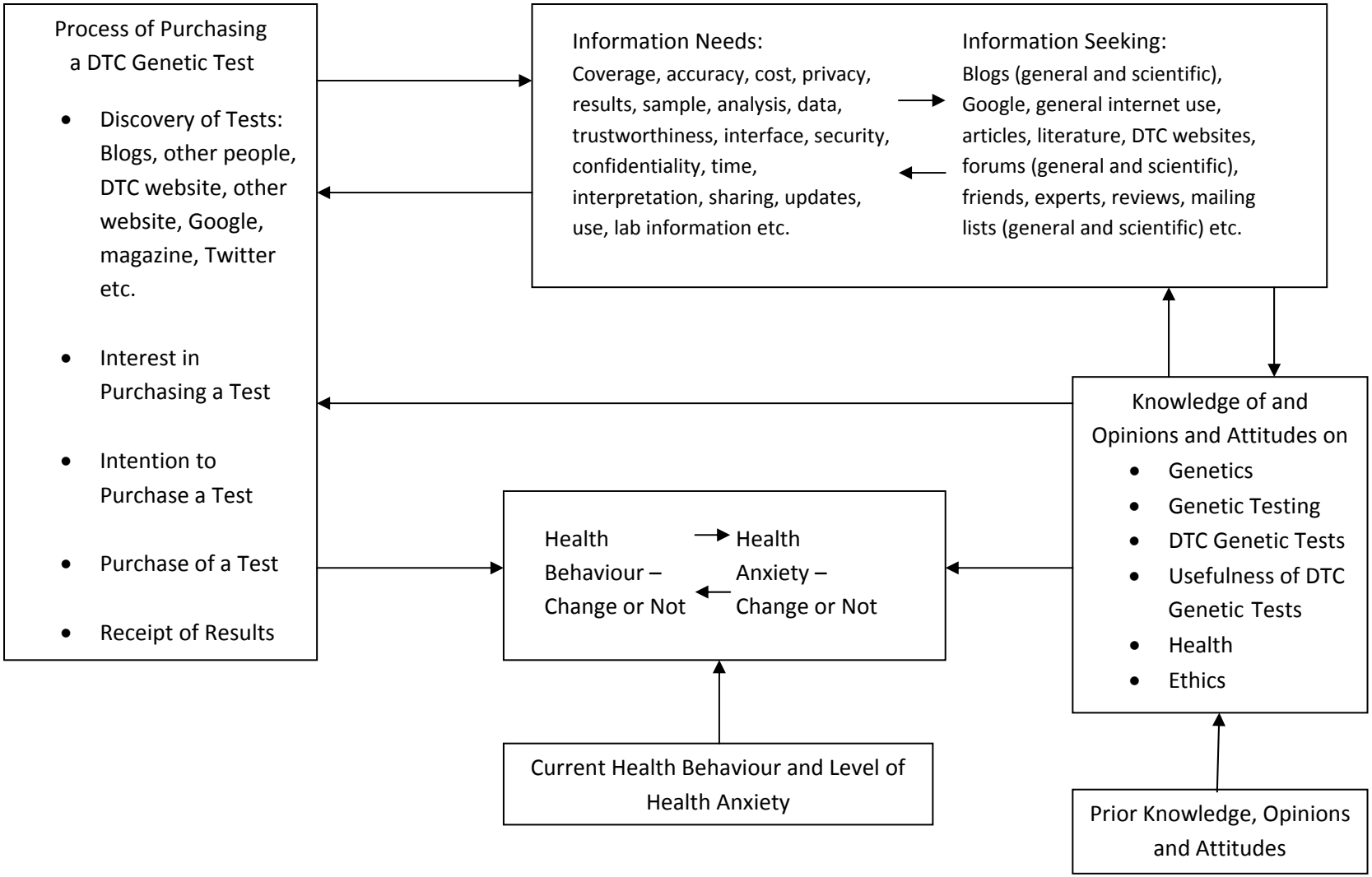


Figure 14 Model of the purchase of a DTC genetic test

The process of purchasing a test, and the receipt of results in particular, are shown as influencing changes to health behaviour and health anxiety; this is an obvious finding of the research. However, other factors can also influence these changes, not least each other. As the model shows, changes in health anxiety can influence changes to health behaviour and vice versa. Changes can also be influenced by current health behaviour and health anxiety; it is clear that the baseline from which an individual begins may affect the changes that they make. Current knowledge and opinions may also influence changes to health behaviour and health anxiety, particularly attitudes towards health and the usefulness of the tests.

8.6.2 Model of the Information-Seeking Behaviour of Consumers of DTC Genetic Tests

The previous model gave a general overview of the process of the purchase of a DTC genetic test. As it touched on many different parts of the process, each component in the model could only be a general summary of the area it represented. Two of the components were, in the context of this research, particularly important, and hence worth a more detailed description: 'information needs' and 'information seeking'. The following model, a model of the information-seeking behaviour of consumers of DTC genetic tests, describes these areas in more depth. As before, the model is neither comprehensive nor universal; it does not seek to cover all of the features of an individual's information-seeking behaviour, neither should it be expected that all consumers will conform to the entirety of the model. Rather, it describes the information needs, information-seeking behaviours and all of the related processes and variables identified in the research. Figure 15 shows this model.

The model begins with 'everyday life information seeking'. This is similar to the component of the same name in Savolainen's model of everyday life information seeking (Savolainen 1995), but less specifically problem orientated. Rather, it refers to all of the information seeking performed by individuals that does not specifically relate to DTC genetic tests, whether it be actively searching for particular task-related information or passively absorbing general information. The information seeking in this component is what leads to an individual's first discovery of DTC genetic tests, and the four sources that survey respondents most commonly mentioned as the source of this discovery are shown in the model.

The next stage of the model is the formation of information needs by the discovery of DTC genetic tests. Information needs (or similar) is a common component of models of information-seeking behaviour (c.f. Wilson 1981; Byström & Järvelin 1995), and in this model describes the information about DTC genetic tests (and related topics) that consumers wish to know. An individual's information needs can be affected both by consumer variables and testing variables; these are

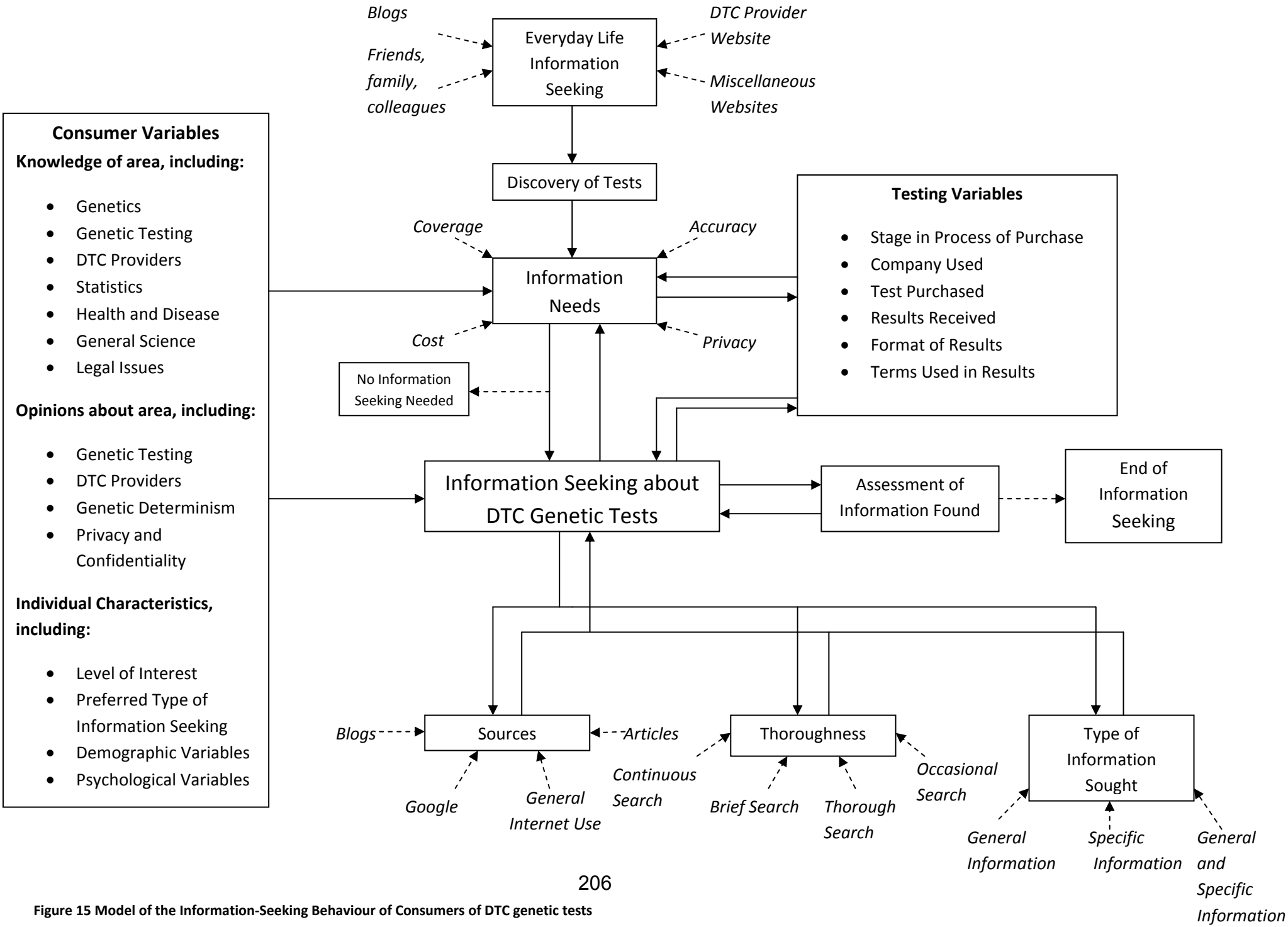


Figure 15 Model of the Information-Seeking Behaviour of Consumers of DTC genetic tests

described in more detail below. The survey and interviews identified a wide range of information needs, the most commonly mentioned of which are shown. In most cases, information needs lead to information seeking. However, the research demonstrated that this is not true in every case, and that some individuals see no need to search for information.

The main component of the model is 'information seeking about DTC genetic tests'. As described above, information seeking is caused by information needs. In turn, the research demonstrated that information seeking may also influence information needs. The model shows that information seeking, along with information needs, can be influenced by two large sets of variables: testing variables and consumer variables. Testing variables are differences in the tests, companies and results which the research has shown to influence some consumers. This can include such variables as the stage of the process of purchasing a test at which an individual is (see Figure 14), which company or test they used, what their results were and how their results were presented. The second component, consumer variables, has also been shown by the research to influence information seeking and information needs. This component has been divided into three different types of variable. The first of these is 'knowledge of area'. As shown in the email interviews, an individual's knowledge about DTC genetic tests and related topics affected both their information need and how they sought information. For example, an individual already knowledgeable about genetic tests may search for more complex information than someone with less background knowledge. Similarly, individuals may be influenced by their opinions of the area, such as the importance of privacy or the trustworthiness of providers, and so this is included as the second type of consumer variable. The third type of consumer variable is individual characteristics; these can be considered as similar to the 'intervening variables' and 'information-seeking behaviour' in Wilson's revised general model of information (Wilson and Walsh 1996). The interviews demonstrated that variables such as demographics and level of interest, and factors such as preferred method of information seeking (e.g. active or passive), influenced consumers' information needs and seeking. Although these are more general than the variables and factors described by Wilson and Walsh (1996) (e.g. their four types of information-seeking behaviour), the relatively small scale of the email interviews meant that, although it can be identified that these factors did influence respondents, it is not possible to describe in detail all of the variables which might do so.

As in many models of information-seeking behaviour there is an inclusion of a feedback loop (c.f. Leckie et al 1996, Wilson et al 1981). This is to demonstrate that individuals assess the information that they have found, and either seek further information or cease information seeking based on the result. However, it should be noted that after cessation information seeking may begin again if

there are further information needs. For example, a consumer may search for information when they first discover the tests, further information when they prepare to order a test and yet further information when they receive their results. Also, the arrow from 'assessment of information found' to 'end of information seeking' is shown as a dotted line in order to indicate that not all individuals cease information seeking; some respondents to the email interviews described it as a continuous process.

The final three components of the model, 'sources', 'thoroughness' and 'type of information sought', are aspects of the information-seeking process. Although these are likely not the only aspects of the process for information seeking in this area, they are those which became apparent during the research. The first, 'sources', refers to the number of sources used by consumers, the different sources they used and issues such as their suitability. The four sources most commonly mentioned by survey respondents are shown, although these are only a small part of a large and extensive list. The second aspect, 'thoroughness', refers to the type of search conducted by respondents e.g. a thorough search or a brief search. Four different options for this aspect are shown, although an individual's actual searching technique would be much more nuanced. The final aspect is 'type of information sought'. During the email interviews it became apparent that many different types of information were sought by consumers, ranging from highly specific, detailed information to general, non-specific information. This is what this aspect refers to, and again, there are several simplified options shown. Although the research showed a large variation between individuals for these three aspects, it should be noted that there may also be variations for different pieces of information sought for by the same individual.

8.7 Summary

This chapter discussed the results of the three studies as a whole. It compared individual results, both with each other and with the literature, and assessed whether or not the research questions had been answered. An ethical analysis of the implications of the research was conducted based on three established bioethical viewpoints. Finally, two models were presented: a model of the purchase of a DTC genetic test and a model of the information-seeking behaviour of consumers of DTC genetic tests.

9 Conclusions

As demonstrated in the discussion, the aim and objectives of the research have been fulfilled and the research questions have been answered. Also, many important contributions to the research area have been made. However, there are some limitations of the research, as well as some further work that should be conducted in the area. This chapter will begin by highlighting the most important contributions that this thesis has made. It will then discuss the implications of the findings in the wider area, identify the limitations of the research and describe any further work that should be conducted. Finally, it will end with a conclusion.

9.1 Contributions

This thesis has made numerous contributions to the research area; the most important are listed below.

- The research involved one of only two studies to have investigated the effects of DTC genetic tests on health behaviour and health anxiety amongst 'real users', and the only one to have compared them to a similar group who differed in receipt of test results.
- The research identified and described mechanisms by which the results from a DTC genetic test can affect consumers' health behaviour and anxiety in a unique level of detail.
- The thesis contains the only research to have examined the informational aspects of DTC genetic tests from the consumers' point of view, including their self-reported information needs, the sources which they used to search for information and their assessment of the information provided by providers' websites.
- The research included the only content analysis of the websites of providers of DTC genetic tests that included criteria generated from consumers' self-reported information needs.
- The research provided data about consumers' experiences with, and opinions about, DTC genetic tests; whilst not unique in the literature these results remain useful.
- The thesis contains the only ethical analysis of DTC genetic tests based on actual findings rather than conjecture.
- The thesis contains the only model of the process of purchasing a DTC genetic test, including both information about and possible effects of the tests, and the only model of the information-seeking behaviour of consumers of DTC genetic tests.
- Parts of the research have been published in an international journal and presented at an international conference.

9.2 Implications of Findings in Wider Area

As described in Chapter 2, DTC genetic tests are a currently-controversial product about which many criticisms and concerns have been expressed. Their future legal position is also far from certain, with many jurisdictions currently pursuing different approaches and others still undecided.

In this context, there are two main implications of the findings. Firstly, that it could be considered unethical for a jurisdiction to ban DTC genetic tests. Although they do not currently have the accuracy and validity to be used in a medical setting, and hence their sale for health purposes could also be considered unethical, the findings point towards a positive effect of the tests on the health behaviour of a minority of consumers. Since the tests are currently available in most jurisdictions, and evidence has not been found for the hypothesised negative effects of the tests, a ban would be more likely to do harm than good.

Secondly, the findings highlight the importance of consumers' information-seeking behaviour. The results have confirmed the findings of previous studies that the information provision on the websites of providers of DTC genetic tests is generally of a low quality. However, the research has demonstrated that, alongside the websites, consumers normally use other sources of information to meet their information needs, and often have further information needs than those identified by professional organisations. This research therefore demonstrates that future research, discussion or action about informed consent or understanding of the tests should not just focus on the information provided by the websites of providers, but also consumers' individual information needs and information-seeking behaviour.

9.3 Limitations

This research has several limitations that should be noted. Firstly, a convenience-sampling technique was used for the survey. As respondents from the survey were contacted for the interviews, this limitation also applies to them. As described in the Research Methodology (section 4.3.2.2), the small proportion of individuals who have purchased a DTC genetic test necessitated a convenience-sampling method in order to contact a large enough sample of consumers whilst remaining independent of the companies that sell the tests, and also to contact a sample of potential consumers. However, the non-random nature of the sample may have added an element of bias to the results, and prevents their generalization to the general population. As respondents were contacted through social media, two further biases also may have been caused. Firstly, the sample will have been restricted to those who have access to the internet. However, since access to the internet is necessary to purchase a DTC genetic test this will likely have not had a large effect.

Secondly, users of social media websites may differ from the average consumer of a DTC genetic test. This is a particular problem with regard to consumers' information seeking, and may have skewed the identified information sources towards blogs and other social media.

A second limitation is with the assessment of the effect of the tests on health behaviour and health anxiety; each of the two methods used had its own limitations. The first method involved asking consumers if their health behaviour or anxiety had changed due to receiving their results. This relied on an accurate memory and assessment by consumers; factors which cannot be guaranteed. It is also possible for it to have been affected by a response bias. The second method compared the health-behaviour scores and health anxiety scores of consumers and potential consumers. Once again, this relied on an accurate assessment by the respondent, but the possibility of recall or response bias was reduced. However, a bias could have been introduced if there was an underlying difference between the two groups, other than receipt of test results. Although these limitations existed, the two methods were used in concert in order to reduce their impact. Also, the significant difference in health-behaviour scores remained significant after weighting for demographic variables and performance of a stepwise multiple regression; reducing the possibility of an underlying difference.

A third limitation is with the content analysis. In order to remove subjective interpretations of the quality of information provided on the websites, the content analysis was designed so that each item was assessed as either covered or not. This meant that the quality or quantity of information was not assessed, and so websites providing differing levels of information about an item may have received the same assessment. However, the content analysis was extremely comprehensive, with a very large number of items assessed. This reduced the potential for bias, as multiple items were assessed for most topics; those websites that provided more information would normally cover more of the items than websites that provided less. It should also be noted that, since the content analysis framework was restricted to items based on either professional recommendations or consumers' answers to the survey questions, it did not have the ability to assess whether websites provided relevant information on the 'cutting edge' of genetic knowledge, such as epigenetics. However, the framework design was a core component of the methodology that allowed for the assessment of the provision of information to be based on what consumers' wished to know and what professionals thought that they should know.

9.4 Further Work

Although this research has made a substantial contribution to the research area, there is still further work to be completed before it can be fully understood.

As stated previously, the research, when combined with previous studies in the literature, suggests that DTC genetic tests have a positive effect on the health behaviours of a minority of consumers. However, research so far cannot prove a causative effect. Future research should focus on doing so, possibly by providing a group of potential consumers with a free test. Although this would be similar to research by Bloss et al (2011b), the use of 'real' potential consumers would be useful in reducing the bias inherent in Bloss's sample selection. Also, an assessment of a much larger and more varied group of health behaviours would be necessary.

The effect of the tests on health anxiety is still far from certain. Although this research, and others in the literature, provide reassurance, a similar study to that suggested in the previous paragraph would be useful for allowing a more thorough picture to emerge. Future research should also investigate the usefulness of different counselling approaches for consumers with different backgrounds e.g. individuals who have poor health behaviour or individuals whose relatives have a known genetic mutation.

The examination of respondents' information behaviour in this research was necessarily exploratory; the area had not been examined by any previous studies and it was important for the results to remain free of any influence. However, this research has now provided data on respondents' information behaviours, and has created a model of consumers' information-seeking behaviour. It should now be possible, therefore, for future research to generate a more comprehensive picture of the area, including a full assessment of the validity of the model.

9.5 Postscript: 23andMe

As described in Chapter 2, in 2012 23andMe became the first provider of DTC genetic tests to file for FDA clearance for a small number of its genetic tests (Allison 2012, p.1027). At the time of submission the FDA had yet to make a decision on this issue. However, in late 2013 the FDA wrote to 23andMe stating that they did not have clearance to market health-related DTC genetic tests and informing them that they must cease to market the product (FDA 2013). Since receiving this letter 23andMe have suspended health reports for all new customers, and now only provide ancestry reports and raw genetic data (23andMe 2013c). However, 23andMe are continuing to seek FDA clearance and in June 2014 submitted a further application to the FDA. This application focuses on

testing for one genetic disorder and is considered as a small step towards full clearance (23andMe 2014).

9.6 Final Conclusion

This thesis has been a timely and useful addition to the research area. At its beginning, research into DTC genetic tests was still at an early stage. The body of literature in this area has now grown to a reasonable size, and this research has contributed both to that and to the understanding of the topic.

A review of the literature identified concerns about the effects of DTC genetic tests on consumers' health behaviour and anxiety and about consumers' ability to give informed consent to the tests with as full awareness as possible of the risks involved. Relevant gaps in the literature were identified, including a need for further assessment of the effects of the tests and a lack of research into the informational aspects of the tests from the consumers' point of view.

In terms of health behaviour, the results and analysis point towards a positive effect of DTC genetic tests on a minority of consumers. Whilst not conclusive, when combined with previous research these results are highly suggestive. Importantly, no negative behavioural effects of the tests were identified. The situation with regard to health anxiety was less clear, with results showing no effect of the tests on the majority and a decrease in health anxiety in a minority. However, several respondents did report an increase in anxiety. In contrast, no difference was found in the current health anxiety of consumers and potential consumers, and so further research into the psychological effects of DTC genetics tests is certainly needed.

Provision of information on providers' websites was found to be poor, with even the website possessing the highest level of coverage still missing many important pieces of information. However, as well as using the websites, consumers were found to have searched for information about the tests independently, using a wide variety of sources. Despite this, most consumers did not search for information on all relevant topics, and so it is likely that concerns about their informed consent are well founded.

Direct to consumer genetic tests are a fascinating and relevant area of study. This thesis is an important contribution to the research area and it is hoped that it will help to illuminate the topic and provide a good basis for further research.

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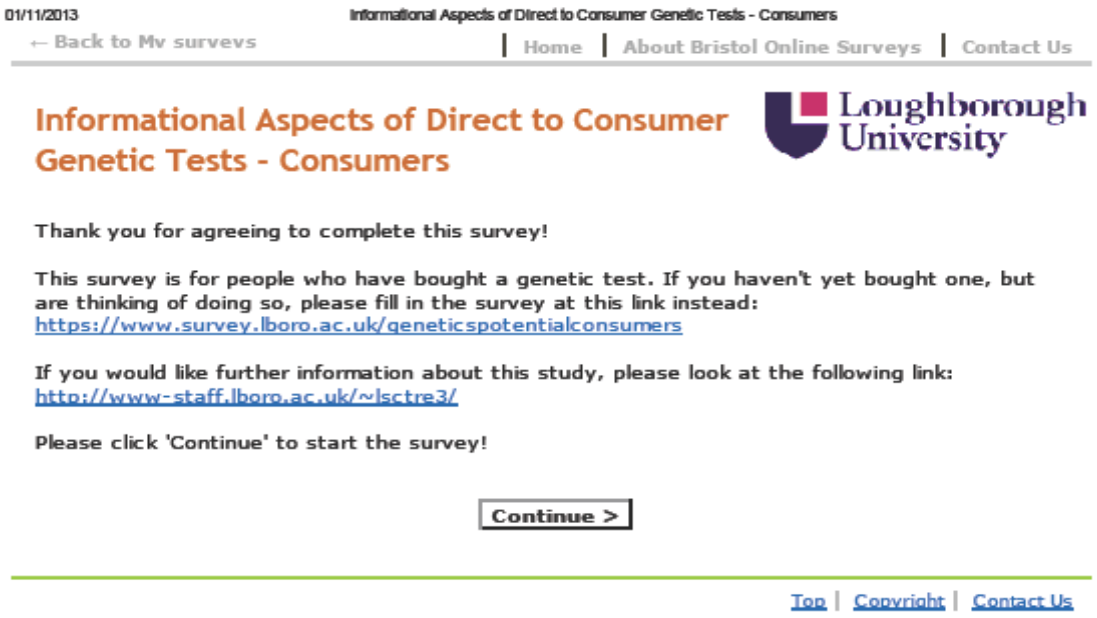
Appendices

Appendix A: Questionnaires

Version for Individuals who had Purchased a DTC Genetic Test (Consumers)



01/11/2013 Informational Aspects of Direct to Consumer Genetic Tests - Consumers

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Informational Aspects of Direct to Consumer Genetic Tests - Consumers

Loughborough University

Thank you for agreeing to complete this survey!

This survey is for people who have bought a genetic test. If you haven't yet bought one, but are thinking of doing so, please fill in the survey at this link instead: https://www.survey.lboro.ac.uk/geneticspotentialconsumers

If you would like further information about this study, please look at the following link: http://www-staff.lboro.ac.uk/~lsctre3/

Please click 'Continue' to start the survey!

Continue >

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https://www.survey.bris.ac.uk/?manifestid=81688&op=preview 1/1

Informational Aspects of Direct to Consumer Genetic Tests - Consumers

Loughborough University

Survey

Please answer as many questions as fully as you can. If there are any questions that you do not feel comfortable answering then please just leave the answer blank.

The Genetic Test

This section is about the genetic test that you have purchased.

1. What is the name of the genetic test that you purchased?

2. Which company did you buy it from?

3. How did you find out about genetic tests?
(select all that apply)

- ☐ Advertising
- ☐ Friends
- ☐ TV (News)
- ☐ TV (Other)
- ☐ Newspaper
- ☐ Magazine
- ☐ The website of a company that sells genetic tests
- ☐ Google
- ☐ Wikipedia
- ☐ Doctor
- ☐ Facebook
- ☐ Twitter
- ☐ Blogs
- ☐ Other internet site
- ☐ Other *(please specify)*:

4. How much did the test cost?

Currency
- ○ UK Pounds
- ○ US Dollars
- ○ Euros

Other *(please specify)*:

5. What were your main reasons for purchasing a genetic test?

6. How long ago did you receive your test results?

- Less than 3 months
- Between 3 and 6 months
- Between 6 and 9 months
- Between 9 and 12 months
- Over a year
- Haven't received them yet

7. Taking an average view of all of the results from your genetic test, how would you interpret your risk of developing a serious disease?

- A significantly higher than average risk of disease
- A slightly higher than average risk of disease
- An average risk of disease
- A slightly lower than average risk of disease
- A significantly lower than average risk of disease
- Other *(please specify)*:

8. Are there any specific details of your test results that you would feel comfortable describing?

- Yes
- No

If yes please describe them

Information Need

9. What information about genetic tests did you want to know before purchasing a test? i.e. information about the test itself, NOT your reason for purchasing the test. (Please list as many answers as you can)

10. Information about genetic tests should be provided on the website of the company from which you purchased the test. How satisfied are you that their website provides sufficient information about the topics you mentioned in answer to the previous question? Please rate your satisfaction on a scale of 1 to 7 (1 least satisfied, 7 most satisfied).

○ 1 ○ 2 ○ 3 ○ 4 ○ 5 ○ 6 ○ 7 ○ N/A

11. After purchasing a genetic test and receiving your results, was there any extra information that you wished to know?

○ Yes
○ No

a. If yes, what was this information?

b. How satisfied are you that their website provides sufficient information about these topics? Please rate your satisfaction on a scale of 1 to 7 (1 least satisfied, 7 most satisfied).

○ 1 ○ 2 ○ 3 ○ 4 ○ 5 ○ 6 ○ 7 ○ N/A

12. Aside from the information provided on the company's website, is there any other information that the company has provided you with?

○ Yes
○ No

If yes, please specify

The Company's Website

This section is about the website of the company from which you purchased the test.

13. Information is provided on the website of the company from which you purchased the genetic test. On a scale of 1 to 7 (1 completely disagree to 7 completely agree), how much do you agree with the following statements?

	1 completely disagree to 7 completely agree						
	1	2	3	4	5	6	7
a. I am generally satisfied with the information provided on the website.	○	○	○	○	○	○	○

b. I had trouble understanding some of the information on the website.							
c. There is adequate information on the website to make a decision about buying a test.							
d. The information on the website appears to be trustworthy.							
e. The information on the website appears to be reliable.							
f. I had to look at other sources to find enough information to make a decision about buying a test.							

Information Searching

This section is about how you looked for information about genetic tests.

14. Please describe briefly what you did when looking for information about genetic tests, including what sources you used, what information you were looking for and how successful you were.

15. Is this typically how you would look for medical/health related information?

- Yes
- No

16. Were you satisified with the information you found?

Yes No

If no, why not?

17. Is this a way of finding information that you would recommend for other people?

- Yes
- No

If no, why not?

18. What source of information would you consider most appropriate for finding out about genetic tests?

Lifestyle Issues

This section will look at certain lifestyle issues, including questions on areas such as diet and exercise. This information is an important part of the study, however, if there are any questions that you feel uncomfortable answering, please feel free to leave them blank.

19. What would you estimate the level of salt is in your diet?

- Lower than or equal to recommended levels
- Higher than recommended levels
- Much higher than recommended levels
- Don't know

20. What would you estimate the level of fat is in your diet?

- Lower than or equal to recommended levels
- Higher than recommended levels
- Much higher than recommended levels
- Don't know

21. What would you estimate the level of fibre is in your diet?

- Higher than or equal to recommended levels
- Lower than recommended levels
- Much lower than recommended levels
- Don't know

22. Do you eat at least five items of fruit or vegetables a day?

- Everyday
- Most days
- Some days
- Rarely
- Don't know

23. Do you drink alcohol (on average)?

- Never
- Less than once a month

- Every 3 or 4 weeks
- Every 1 or 2 weeks
- 1 or 2 days a week
- 3 or 4 days a week
- 5 or 6 days a week
- Everyday

If you drink one or more days a week, approximately how much alcohol do you drink per session?

24. Do you smoke cigarretes?

- Never
- Less than 1 per day
- Between 1 and 10 per day
- Between 11 and 20 per day
- Over 20 per day

25. Do you smoke cigars or a pipe?

- Never
- Very occasionally
- Occasionally
- Often

26. Approximately how much vigerous physical activity do you do per week? Examples of vigerous physical activity include running, jogging, racewalking, cycling at 10 mph or more, swimming laps, playing singles tennis, playing football (soccer), heavy gardening, walking with a heavy backpack, lifting weights, aerobic dancing and exercise classes.

	Length of Time (minutes)						Number of days per week						
	None	1 to 15	16 to 30	31 to 45	46 to 60	Over 60	1	2	3	4	5	6	7
a. Vigerous Physcial Activity													

27. Approximately how much moderate physical activity do you do per week? Examples of moderate physical activity include going for a brisk (at least 3 mph) or long walk, water aerobics, cycling slower than 10 mph, playing doubles tennis, ballroom dancing, general gardening and carrying heavy bags back from the shops.

Length of Time (minutes)						Number of days per week						
None	1 to	16 to	31 to	46 to	Over 60	1	2	3	4	5	6	7

		15	30	45	60								
a. Moderate Physical Activity													

28. Approximately how much light physical activity do you do per week? Examples of light physical activity include going for a short walk, light gardening, DIY, light housework and using the stairs.

	Length of Time (minutes)						Number of days per week						
	None	1 to 15	16 to 30	31 to 45	46 to 60	Over 60	1	2	3	4	5	6	7
a. Light Physical Activity													

29. On a scale of 1 to 10 (1 least anxious, 10 most anxious), how anxious do you normally feel about:

	1	2	3	4	5	6	7	8	9	10
a. The state of your health										
b. The possibility of developing a serious disease										

30. Did your health-related behaviour change at all, either positively or negatively, due to recieving your test results?

- Yes
- No

If yes, how did it change and have these changes lasted until now?

31. Did your level of health-related anxiety change at all due to receiving your test results?

- Yes
- No

If yes, how did it change and have these changes lasted until now?

About You

32. What is your gender?

- Male
- Female

33. What is your age?

- 18-29
- 30-44
- 45-60
- 60+

34. Please select your country of birth

Select an answer

If you selected Other, please specify:

35. Do you live in a different country to that of your birth?

- Yes
- No

a. If yes, where do you live now?

Select an answer

b. How long have you lived there?

36. How would you describe your ethnicity?

Please note that this question is entirely voluntary; you don't have to answer it if you would prefer not to.

37. What is the highest level of education that you have completed?

Employment Status

The purpose of this section is to determine your employment status. The questions refer to your current main job, or (if you are not working now) to your last main job.

38. Do (did) you work as an employee or are (were) you self-employed?

- Employee
- Self-employed with employees
- Self-employed / freelance without employees
- Other *(please specify)*:

a. If your answer was "employee", how many people work (worked) for your employer at the place where you work (worked)?

- 1 to 24
- 25 or more

b. If your answer was "self-employed with employees", how many people do (did) you employ?

- 1 to 24
- 25 or more

39. Do (did) you supervise any other employees? More Info

- Yes
- No
- N/A

40. Please select which of the following best describes the sort of work you do (did).

- Modern professional occupations (e.g. teacher, nurse, physiotherapist, social worker, welfare officer, artist, musician, software designer)
- Clerical and intermediate occupations (e.g. secretary,personal assistant,clerical worker,office clerk, call centre agent, nursing auxiliary, nursery nurse)
- Senior managers or administrators (usually responsible for planning, organising and co-ordinating work and for finance, e.g. finance manager, chief executive)
- Technical and craft occupations (e.g. motor mechanic, fitter, inspector, plumber, printer, tool maker, electrician, gardener, train driver)
- Semi-routine manual and service occupations (e.g. postal worker, machine operative, security guard, caretaker, farm worker, catering assistant, receptionist, sales assistant)
- Routine manual and service occupations (e.g. HGV driver, van driver, cleaner, porter, packer, sewing machinist, messenger, labourer, waiter / waitress, bar staff)
- Middle or junior managers (e.g. office manager, retail manager, bank manager, restaurant manager, warehouse manager, bar manager)
- Traditional professional occupations (e.g. accountant, solicitor, medical practitioner, scientist, civil / mechanical engineer)
- Other *(please specify)*:

Username

41. If you think you may want to withdraw from this study at some point in the future, or otherwise contact us about your answers, please enter a username. As this is an anonymous question, this will allow us to identify your answers. This is entirely voluntary.

Contact

42. If you would be happy to be contacted about participating in further research in this area, please enter your email address. This is entirely voluntary.

Continue >

Survey testing only

Check Answers & Continue >

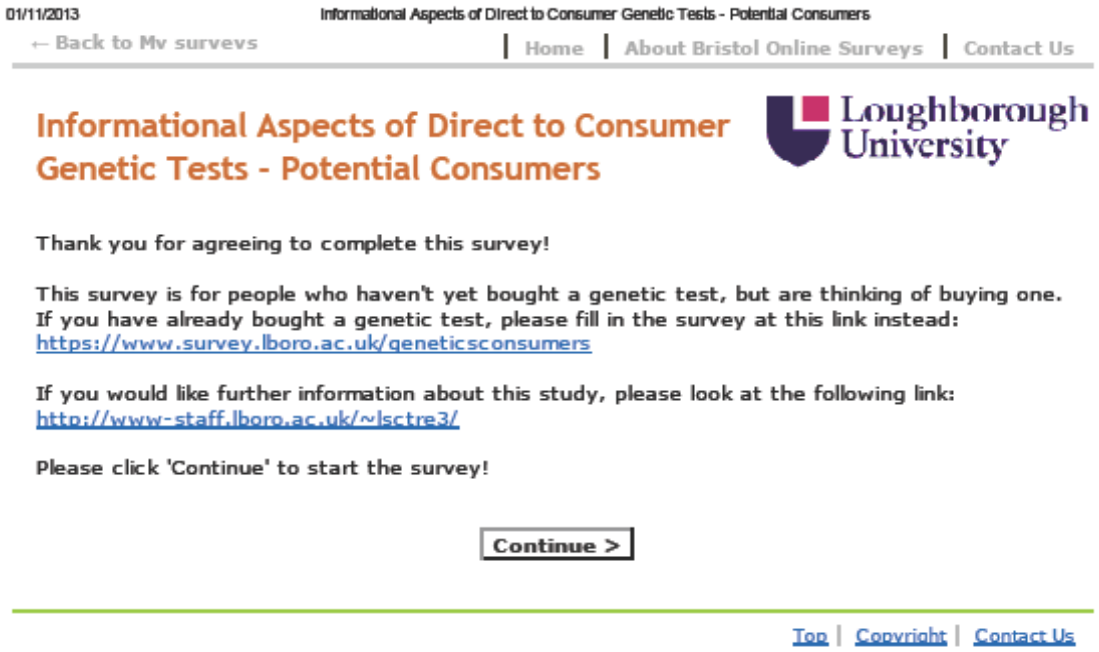
Informational Aspects of Direct to Consumer Genetic Tests - Consumers

Loughborough University

Final Page

Thank you for completing this survey!

Version for Individuals who were Considering Purchasing a DTC Genetic Test (Potential Consumers)



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Informational Aspects of Direct to Consumer Genetic Tests - Potential Consumers

Loughborough University

Thank you for agreeing to complete this survey!

This survey is for people who haven't yet bought a genetic test, but are thinking of buying one. If you have already bought a genetic test, please fill in the survey at this link instead: https://www.survey.lboro.ac.uk/geneticsconsumers

If you would like further information about this study, please look at the following link: http://www-staff.lboro.ac.uk/~lsctre3/

Please click 'Continue' to start the survey!

Continue >

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Informational Aspects of Direct to Consumer Genetic Tests - Potential Consumers

Loughborough University

Survey - Page 1

Please answer as many questions as fully as you can. If there are any questions that you do not feel comfortable answering then please just leave the answer blank.

The Genetic Test

This section is about the genetic test that you are thinking of purchasing.

1. On a scale of 1 to 10 (1 least seriously, 10 most seriously), how seriously are you thinking of purchasing a genetic test?

○ 1 ○ 2 ○ 3 ○ 4 ○ 5 ○ 6 ○ 7 ○ 8 ○ 9 ○ 10

2. Is there any particular company that you're thinking of purchasing a genetic test from?

○ Yes
○ No

If yes, which company?

3. Is there any particular genetic test that you're thinking of purchasing?

○ Yes
○ No

a. If yes, which test?

b. How much does it cost?

c. Currency

○ UK Pounds
○ US Dollars
○ Euros
○ Other *(please specify)*:

4. How did you find out about genetic tests?
(select all that apply)

☐ Advertising
☐ Friends

- ☐ TV (News)
- ☐ TV (Other)
- ☐ Newspaper
- ☐ Magazine
- ☐ The website of a company that sells genetic tests
- ☐ Google
- ☐ Wikipedia
- ☐ Doctor
- ☐ Facebook
- ☐ Twitter
- ☐ Blogs
- ☐ Other internet site
- ☐ Other *(please specify)*:

5. What are your main reasons for wanting to purchase a genetic test?

Information Need

6. What information about genetic tests do you want to know before purchasing a test? i.e. information about the test itself, NOT your reason for purchasing the test. (Please list as many answers as you can)

7. Information about genetic tests should be provided on the websites of the companies that sell them. If there is a particular company from which you are considering purchasing a test, how satisfied are you that their website provides sufficient information about the topics you mentioned in answer to the previous question? Please rate your satisfaction on a scale of 1 to 7 (1 least satisfied, 7 most satisfied).

○ 1 ○ 2 ○ 3 ○ 4 ○ 5 ○ 6 ○ 7

The Company's Website

This section is about the website of the company from which you purchased the test.

8. Information is provided on the websites of companies that sell genetic tests. If there is a particular company from which you are considering purchasing a test, on a scale of 1 to 7 (1 completely disagree to 7 completely agree), how much do you agree with the following statements?

	1 completely disagree to 7 completely agree

	1	2	3	4	5	6	7
a. I am generally satisfied with the information provided on the website.	○	○	○	○	○	○	○
b. I had trouble understanding some of the information on the website.	○	○	○	○	○	○	○
c. There is adequate information on the website to make a decision about buying a test.	○	○	○	○	○	○	○
d. The information on the website appears to be trustworthy.	○	○	○	○	○	○	○
e. The information on the website appears to be reliable.	○	○	○	○	○	○	○
f. I have had to look at other sources to find enough information to help me make a decision on whether or not to buy a genetic test.	○	○	○	○	○	○	○

Information Searching

This section is about how you looked for information about genetic tests.

9. Please describe briefly what you did when looking for information about genetic tests, including what sources you used, what information you were looking for and how successful you were.

10. Is this typically how you would look for medical/health related information?

- ○ Yes
- ○ No

11. Were you satisified with the information you found?

- ○ Yes
- ○ No

If no, why not?

12. Is this a way of finding information that you would recommend for other people?

- Yes
- No

If no, why not?

13. What source of information would you consider most appropriate for finding out about genetic tests?

Lifestyle Issues

This section will look at certain lifestyle issues, including questions on areas such as diet and exercise. This information is an important part of the study, however, if there are any questions that you feel uncomfortable answering, please feel free to leave them blank.

14. What would you estimate the level of salt is in your diet?

- Lower than or equal to recommended levels
- Higher than recommended levels
- Much higher than recommended levels
- Don't know

15. What would you estimate the level of fat is in your diet?

- Lower than or equal to recommended levels
- Higher than recommended levels
- Much higher than recommended levels
- Don't know

16. What would you estimate the level of fibre is in your diet?

- Higher than or equal to recommended levels
- Lower than recommended levels
- Much lower than recommended levels
- Don't know

17. Do you eat at least five items of fruit or vegetables a day?

- Everyday
- Most days
- Some days
- Rarely
- Don't know

18. Do you drink alcohol (on average)?

- Never
- Less than once a month
- Every 3 or 4 weeks
- Every 1 or 2 weeks
- 1 or 2 days a week
- 3 or 4 days a week
- 5 or 6 days a week
- Everyday

If you drink one or more days a week, approximately how much alcohol do you drink per session?

19. Do you smoke cigarretes?

- Never
- Less than 1 per day
- Between 1 and 10 per day
- Between 11 and 20 per day
- Over 20 per day

20. Do you smoke cigars or a pipe?

- Never
- Very occasionally
- Occasionally
- Often

21. Approximately how much vigerous physical activity do you do per week? Examples of vigerous physical activity include running, jogging, racewalking, cycling at 10 mph or more, swimming laps, playing singles tennis, heavy gardening, walking with a heavy backpack, lifting weights, aerobic dancing and exercise classes.

	Length of Time (minutes)						Number of days per week						
	None	1 to 15	16 to 30	31 to 45	46 to 60	Over 60	1	2	3	4	5	6	7
a. Vigerous Physcial Activity													

22. Approximately how much moderate physical activity do you do per week? Examples of moderate physical activity include going for a brisk (at least 3 mph) or long walk, water aerobics, cycling slower than 10 mph, playing doubles tennis, ballroom dancing, general gardening and carrying heavy bags back from the shops.

	Length of Time (minutes)						Number of days per week						

	None	1 to 15	16 to 30	31 to 45	46 to 60	Over 60	1	2	3	4	5	6	7
a. Moderate Physical Activity	○	○	○	○	○	○	○	○	○	○	○	○	○

23. Approximately how much light physical activity do you do per week? Examples of light physical activity include going for a short walk, light gardening, DIY, light housework and using the stairs.

	Length of Time (minutes)						Number of days per week						
	None	1 to 15	16 to 30	31 to 45	46 to 60	Over 60	1	2	3	4	5	6	7
a. Light Physical Activity	○	○	○	○	○	○	○	○	○	○	○	○	○

24. On a scale of 1 to 10 (1 least anxious, 10 most anxious), how anxious do you normally feel about:

	1	2	3	4	5	6	7	8	9	10
a. The state of your health	○	○	○	○	○	○	○	○	○	○
b. The possibility of developing a serious disease	○	○	○	○	○	○	○	○	○	○

25. If you buy a genetic test and the results show a signficant increased or decreased risk of serious disease, do you think that this will change your health-related behaviour?

○ Yes
○ No

Please give details

26. If you buy a genetic test and the results are neutral (i.e. show no overall increased or decreased risk of serious disease) do you think that this will change your health-related behaviour?

○ Yes
○ No

Please give details

27. If you buy a genetic test and the results show a signficant increased or decreased risk of serious disease, do you think that this will change your health-related anxiety?

- Yes
- No

Please give details

28. If you buy a genetic test and the results are neutral (i.e. show no overall increased or decreased risk of serious disease), do you think that this will change your health-related anxiety?

- Yes
- No

Please give details

About You

29. What is your gender?

- Male
- Female

30. What is your age?

- 18-29
- 30-44
- 45-60
- 60+

31. Please select your country of birth

Select an answer

If you selected Other, please specify:

32. Do you live in a different country to that of your birth?

- Yes
- No

a. If yes, where do you now live?

Select an answer

b. How long have you lived there?

33. How would you describe your ethnicity?

Please note that this question is entirely voluntary; you don't have to answer it if you would prefer not to.

34. What is the highest level of education that you have completed?

Employment Status

The purpose of this section is to determine your employment status. The questions refer to your current main job, or (if you are not working now) to your last main job.

35. Do (did) you work as an employee or are (were) you self-employed?

- Employee
- Self-employed with employees
- Self-employed / freelance without employees
- Other *(please specify)*:

a. If your answer was "employee", how many people work (worked) for your employer at the place where you work (worked)?

- 1 to 24
- 25 or more

b. If your answer was "self-employed with employees", how many people do (did) you employ?

- 1 to 24
- 25 or more

36. Do (did) you supervise any other employees? More Info

- Yes
- No
- N/A

37. Please select which of the following best describes the sort of work you do (did).

- Modern professional occupations (e.g. teacher, nurse, physiotherapist, social worker, welfare officer, artist, musician, software designer)
- Clerical and intermediate occupations (e.g. secretary,personal assistant,clerical worker,office clerk, call centre agent, nursing auxiliary, nursery nurse)
- Senior managers or administrators (usually responsible for planning, organising and co-ordinating work and for finance, e.g. finance manager, chief executive)
- Technical and craft occupations (e.g. motor mechanic, fitter, inspector, plumber, printer, tool maker, electrician, gardener, train driver)
- Semi-routine manual and service occupations (e.g. postal worker, machine operative, security guard, caretaker, farm worker, catering assistant, receptionist, sales assistant)
- Routine manual and service occupations (e.g. HGV driver, van driver, cleaner, porter, packer, sewing machinist, messenger, labourer, waiter / waitress, bar staff)
- Middle or junior managers (e.g. office manager, retail manager, bank manager, restaurant manager, warehouse manager, bar manager)
- Traditional professional occupations (e.g. accountant, solicitor, medical practitioner, scientist, civil / mechanical engineer)
- Other *(please specify)*:

Username

38. If you think you may want to withdraw from this study at some point in the future, or otherwise contact us about your answers, please enter a username. As this is an anonymous question, this will allow us to identify your answers. This is entirely voluntary.

39. If you would be happy to be contacted about participating in further research in this area, please enter your email address. This is entirely voluntary.

Continue >

Survey testing only

Check Answers & Continue >

Informational Aspects of Direct to Consumer Genetic Tests - Potential Consumers

Loughborough University

Final Page

Thank you for completing this survey!

Appendix B: Questionnaire Pilot

Comments on Consumers' Questionnaire

Either use UK for all UK places of birth or England, Wales, Ireland, Scotland. Having both options is confusing.

There are quite a few questions in the survey, but they are all on one page. I have had more success in the past by splitting questions across multiple pages, maybe 10 questions on each, because then people don't feel quite so daunted by the amount that they have to fill in. Otherwise people look at the whole survey and get a bit scared. Sometimes survey engines allow people to fill in a page at a time, and then return at a later date - not sure if yours does or not.

I tried not providing answers to some important questions, but no error messages were generated - normally, there is a way with survey software to mark questions as being mandatory or not. For some questions, you probably would want to do this, otherwise you might get lots of answers that miss key data (like age, sex etc). Especially near the bottom of the survey where people might be getting a bit bored.

Should you have a link to a page of information describing your research? or maybe not, not quite sure. Maybe depends on whether someone read something first before clicking on the link.

I found question 8 a bit confusing (8. Was there any information that was important to your decision to buy a genetic test?). Maybe there is a slightly different way of wording this? e.g. was there any information that influenced your purchase of a genetic test?

Q5 - is this questionnaire only for people who have had results then? Q6 - asking people to interpret stuff, bit asking them about the test properly - is this right? 'Information need' and 'information topics' these are researcher terms, not user terms. Need rewriting. Perhaps do as you have in other sections and say 'this section is about....' to make things clearer. Q10 - end bit is odd - needs rewording to make more clear. Q11 - information topic again. Q13 - trustworthy and reliable? Q13 f - are you asking this question about info from a website about tests in general, their test, the results or what? In 'f' you specify but not in the other answers. You want them to type a lot - this may limit answers as people may find this time consuming (much easier to press a button!). How long do you think this questionnaire will take? Q14 is very broad and you're asking a lot from one question. Can it be broken down a bit further without repeating anything asked before? Perhaps 'company website'

section should be after 'information searching' section? Just seems to flow better as a reflection of what they may have done. Lifestyle issues - where is the warning that these questions are coming? They seem intrusive and very personal. Not clear on the relevance of some of them. How are people supposed to know what 'average' is? I wouldn't like to answer some of these. Who smokes a pipe these days (is it worth a separate question?)? Cigarettes is spelt wrong. Demographics and Socioeconomic status - researcher terms so need rewriting. Overall I think you need to try to think like a respondent for some of this. Would you like to answer all these questions? Are all the terms and the language and instructions clear? What's the relevance of some of this? Q26 - intensity? who thinks like that?

Problems Noticed on Consumers' Questionnaire

For question 8 (Was there any information that was important to your decision to buy a genetic test?) one person answered 'yes' then specified 'curiosity'.

Comments on Potential Consumers' Questionnaire

It's fine

With the likert scale questions, it would be more useful to display the options (for example 1 completely disagree to 7 completely agree) above the actual scale. The scale might be a little easier to interpret this way. - Question 20 is difficult to answer. Could perhaps make it more simple.

The move from section 6 to 7 is slightly confusing as you go from 'satisfaction' to 'agreeing/not agreeing'. Apart from that it's fine.

Q-3 did you want to know what condition the test was for? Q-6 no space for not applicable, I first answered that I did not know what company I would use but then changed it to answer the survey. Q-7 No place to say that I have not purchased the test, someone may be thinking of it, but not yet done it for some reason. Q-20 A description of the intensity would be useful. Also, I tried to check 15 mins of moderate for 5 days a week and over 60 mins of moderate for 1 day a week to reflect my exercise of walking 15mins each weekday, and several hours of allotment work at the weekend, but I couldn't do that.

It was very clear

I'd be careful with your 'yes-no' answers though, as you ask to explain the yes but not the no (or vice versa)-- make sure you don't jump to quickly and assume cause they said no it means X.
-- Might want to re-word question 6 (second sentence probably not relevant).

-- Question 8 (re word as Please describe briefly). Some people could find it rude to say 'short paragraph' as they might not give you as much information as you'd like.

-- For questions 13, 14 & 15 it would be useful to give an example of the 'recommended levels' -- I didn't know what they were till I checked.

-- Question 22, 23, 24 & 25- part b, what details are you looking for? Hit them with a question or ask for a summary on what information you need. What details do you need?

-- Is it possible to re-arrange question 31 so after the answer you link, e.g. Employee (Go to 31a) Self employed with employees (Go to 31b), then take out the first part and just write the question, e.g. a. How many people work..., b. How many people do/did you employ?

-- Question 33 - Take out all the times you write 'occupations' in the answers and just write occupations in the title (e.g. what best describes your occupation). And instead of using 'or' in the answers use /. And take out 'such as' and use e.g.

I have just a few comments, regarding the questions 7, and 22 to 25.

As I looked to question 7, I felt there was a certain redundancy... I mean, if "I am generally satisfied with the information provided on the website" (a.), of course I have not "had trouble understanding some of the information on the website" (b.)... And then we have the statement "There is adequate information on the website to make a decision about buying a test" (c.), which seems to be kind of repetitive. And finally, is not the word 'trustworthy' (d.) a synonym of 'reliable' (e.)? If there is technical difference between those words, it is good to explain it to the participants. Otherwise, it seems duplicate again.

Regarding the Questions 22 to 25, they seem to address very similar issues. Isn't there a way of merging into one or maybe two questions, instead of 4?

All in all, I think the questionnaire was well designed and can be easily understood

Problems Noticed on Potential Consumers Questionnaire

One person just wrote 'yes' in the text box for questions 2 and 3 (Is there any particular company that you're thinking of purchasing a genetic test from? and Is there any particular genetic test that you're thinking of purchasing?) rather than putting in the names of the company and test.

Although her current employment status is a PhD student, one respondent filled out the employment status section for her last job.

Appendix C Information Pages

Survey Participant Information Page

The Informational Aspects of Genetic Tests

Thank you for your interest in this survey!

My name is Corin Egglestone, and I'm a PhD student in the Department of Information Science at Loughborough University in the UK. For my PhD studies I'm doing research into the informational aspects of genetic tests. The first part of this research is a survey of people who have either bought or are thinking of buying a genetic test.

There are different types of genetic tests, and this survey is focused on those tests that include an estimate of your risk for various different diseases, rather than tests that just focus on ancestry, personal traits or a single disease. If you have bought such a test or are thinking of buying one, would you be willing to fill in this survey?

The survey is anonymous and confidential. There is no obligation to take part, and you can withdraw from the study at any time. It should only take about 10-15 minutes, and I'd be really grateful if you'd be willing to fill it in.

If you wish to contact me please email me at C.T.R.Egglestone3@lboro.ac.uk or look at my University Webpage

For extra information about this survey, please click here

To fill in the survey, please:

Click here if you have bought a genetic test. (If you have bought a test but haven't recieved your results yet, please still follow this link and just ignore any questions you can't answer).

Click here if you are thinking of buying a genetic test.

Thanks!

Survey Extra Information Page

Extra Information for Participants

This survey is being conducted by Corin Egglestone as part of his PhD studies, and supervised by Dr Ann O'Brien and Professor Anne Morris in the Department of Information Science at Loughborough University in the UK.

The purpose of this survey is to investigate the informational aspects of genetic tests. This covers both the information provided for consumers about genetic tests, and any effects of the information generated by genetic tests. There are different types of genetic tests, and this survey is focused on those tests that include an estimate of your risk for various different diseases, rather than tests that just focus on ancestry, personal traits or a single disease. You are being asked to complete this survey because you have either bought such a test or are thinking of buying one.

In the survey you will be asked questions about your opinion on the information that is provided/should be provided on the websites of the companies that sell genetic tests, the way that you looked for information about genetic tests and issues to do with your lifestyle (e.g. how much exercise you do). There will also be some demographic questions, questions about your occupation and about the genetic test you bought or are thinking of buying. The survey should take less than 15 minutes to fill in.

This survey is anonymous and confidential. All handling and storage of your data will comply with the UK Data Protection Act 1998. Raw Data will be stored electronically on password protected computer accounts. Any (voluntarily given) email addresses will be stored seperately to the rest of the raw data, and no data that could be used to identify participants will be included in any publication. Only the study investigator, his supervisors and his examiners will have access to any piece of raw data. The data will only be used for the investigator's PhD projects and any academic publications arising from the research.

You are under no obligation to participate in this study.

You are free to withdraw your results from the study at any time, and can do so by contacting Corin on the email address provided below. Please note, since this is an anonymous questionnaire, you will only be able to withdraw from the study if you specify a username in the survey and quote this in any communications. If you ask to withdraw from the study you will not be asked to expain your reason/s for withdrawing.

If you wish to contact us please email Corin at C.T.R.Egglestone3@lboro.ac.uk or look at Corin's University Webpage

To fill in the survey, please:

Click here if you have bought a genetic test. (If you have bought a test but haven't recieved your results yet, please still follow this link and just ignore any questions you can't answer).

Click here if you are thinking of buying a genetic test.

If you are unhappy with how the research has been conducted, or wish to report anything relating to research misconduct, please click here.

Appendix D Email Interview Contacting Respondents

Introduction Email

Hi

Last year you filled in a survey about genetic testing as part of my PhD research at Loughborough University in the UK. I received a good number of replies and have finished analysing all of the information, and so I'm now moving onto my next project. In the survey you indicated that you would be happy to be contacted about further research, and so I was wondering if you would be willing to participate in it.

There were some interesting issues raised in the survey, and my aim is to look at these in more depth. I plan to do this by asking some further questions over email, with several emails sent back and forth in an email conversation.

The email conversation would be completely confidential, with no obligation to take part, and you would be able to withdraw from the study or call a halt to the conversation at any time. Although several emails will be sent, each one should only take a couple of minutes of your time, with no rush to send emails to any particular schedule. Further information about the study can be found at http://www-staff.lboro.ac.uk/~lsctre3/information2.html

Please let me know if you'd be willing to participate in this study or not.

Thanks

Corin Egglestone

http://www.lboro.ac.uk/departments/dis/research/PhDstudents/CEgglestone.html

Participant Information Page

Information for Participants

This study is being conducted by Corin Egglestone as part of his PhD studies, and supervised by Dr Ann O'Brien and Professor Anne Morris in the Department of Information Science at Loughborough University in the UK.

After a successful survey last year, the purpose of this study is to further investigate the informational aspects of genetic tests, examining several issues raised in the survey in detail. Similarly to the survey, these issues are to do with both the information provided for consumers about genetic tests, and any effects of the information generated by genetic tests.

The study will take the form of email conversations. Corin will email you with a question, and your reply will be used as the basis for further questions. Although several emails will be sent back and forth, each one should only take a couple of minutes of your time, and the process will be spread out over several weeks.

All data will be anonymised, with no contact details or data that could be used to identify participants included in any report or publication. Some of your (anonymised) answers may be quoted in the investigator's PhD thesis and/or any academic publications arising from the research, unless you specifically request for this not to happen. All handling and storage of your data will comply with the UK Data Protection Act 1998. Raw Data will be stored electronically on password protected computer accounts, with email addresses stored in a seperate file to the rest of the raw data. The data will only be used for the investigator's PhD projects and any academic publications arising from the research.

You are under no obligation to participate in this study. You are free to withdraw your results from the study at any time, and can do so by informing Corin via email. You may refuse to answer any question or call a halt to your participation in the study at any time. You will not be asked to expain your reason/s for withdrawing, halting your particpation or refusing to answer a question.

If you wish to contact us please email Corin at C.T.R.Egglestone3@lboro.ac.uk or look at Corin's University Webpage

If you are unhappy with how the research has been conducted, or wish to report anything relating to research misconduct, please click here.

Appendix E Generation of Items for Content Analysis

Items Generated from Survey Responses

Question	Response	Distilled
[Consumers questionnaire] 9. What information about genetic tests did you want to know before purchasing a test? i.e. information about the test itself, NOT your reason for purchasing the test. (Please list as many answers as you can)	who the testing company might share results with	Third parties with access to results
	Cost #SNPs tested Chip used Lab certification Ability to share	Cost of test Chip used Certification of Laboratory Ability to share results
	I wanted to be able to match results with other persons taking the test.	Ability to match results with other users
	How much cost How sample collected What information would be analysed How accurate were tests	Cost How sample collected What information analysed Accuracy of tests
	I wanted to know about confidentiality, especially concerning health insurance. If disease risk showed something that could require expensive treatment there is a chance that I could lose health insurance or have the cost increase dramatically. How accurate is the information - meaning are the results so general that they are not of any use.	Confidentiality – general Confidentiality – health insurance Accuracy of tests
	1)The reliability aka how accurate and rate of errors 2) Did they give information as to the sample size.	Accuracy of tests Reliability of tests Sample size
	I had reservations due to watching movies that cloned people I was very interested in the details around how insurance companies could treat this information if they had access to it.	Risks – insurance companies
	Privacy policy and how data would be used by the company	Privacy policy Use of data by company
		How test administered

	how it was administered, is the information shared with insurance companies If I would have access to the raw data. What disease, traits, carrier it was testing How much, how reliable The amount of data that can be obtained, number and type of diseases covered by the test, etc Security was very important for me. I did not want the information to leak so parts of governments would have copy of my genome and make a super-fat-soldier. Jokes aside, I was worried about security. I was worried about misinterpretation of result. But 23 and Me did a good job (well, at least no clones yet). How private my data would be kept, the security of that data How many markers, raw access to data, depth of analytics how many snps and on what platform do they test Cost, number of SNP's What technology was used? How can I get my raw data? data coverage, updates What diseases it would test for. Methods used. Reliability of	Access to information by insurance companies Consumer access to raw data What the test covers – disease/traits/carrier testing Cost of test Reliability of test Amount of data that can be obtained What the test covers - diseases Security issues Accuracy of interpretation Privacy issues Security of data How many markers looked at Consumer access to raw data Depth of Analysis Number of SNPs tested Platform used for testing Cost of test Number of SNPs tested Technology used Consumer access to raw data Coverage of test Updates to results What the test covers – diseases Methods used – general Reliability of results

	results. How detailed the ancestry would be. How long to wait for results. Cost. how the company was going to potentially store, use, and share the information Which are the SNPs, what are the prevention strategies available How comprehensive. Quality of user interface. Ability to move data to other systems Privacy Transparency of process Scientific credibility Accuracy of test Privacy of sequenced data Regions of the genome tested and how they were tested. reliability, who is the company selling the test, is the company trustworthy How the test process works Level of genomic coverage How reliable were the tests. What was the repeatability of the results when done using different testing methods by different companies. I have had my Y DNA tested by three companies and all were in agreement on the counts at the same markers even though each company used a different test method. cost and genes/SNPs tested Specifically, i wanted to know	Time taken Cost of test Storage of data How company uses data Third party access to data SNPs tested Possible prevention strategies Comprehensiveness of test Quality of user interface Consumer access to raw data Privacy issues Transparency of process Scientific credibility of results Accuracy of test Privacy issues What test covers – genome Methods of DNA testing Reliability of test Company details Test process What test covers – genome Reliability of tests Similarity of results between different companies Cost of test SNPs tested Genes tested Consumer access to raw data If company goes out of business

	a) that I could download all snp data, and b) that if 23&me were to go out of business, that the data they had on file would be destroyed Price Comprehensiveness (e.g. how many SNPs analyzed) Company's good reputation Whether the laboratory is certified / follows established testing procedures how to take the DNA sample What types of information I might receive. How accurate they were. If the information could be used by private companies or research companies. Any downfalls to taking the test. I wanted to know if I could download the raw SNP data and what info I'd have access to. data quality ancestry, disease risk, interest in how info was delivered whether i had the ability to download raw data, what it could and couldn't test for test accuracy Price privacy # of SNPs sequenced what the data would be used for I wanted to know whether they would actually be informative about disease.	– fate of data Cost of test Number of SNPs analysed Certification of laboratory Whether laboratory follows established testing procedures Method of taking DNA sample Information provided Accuracy of tests Third party access to data Downfalls of taking the test# Consumer access to raw data Information provided Quality of data How information is provided Consumer access to raw data What could be tested for What couldn't be tested for Accuracy of test Cost of test Privacy issues Number of SNPs sequenced How company uses data Usefulness of test – in relation to disease Accuracy of test

	Data accuracy, number of variants tested The only thing I really cared about was coverage. I would love to sequence my whole genome, but that's still way too expensive. Given that the US FDA may end up crippling the DTC genetic testing industry I figured I would go with something like the 23andMe service for now that uses a pretty dense SNP array for testing. I wanted to know how many SNPs were tested and the likelyhood that these SNPs would be found significant in disease now and in the future as opposed to SNPs not on the chip or on different chips. I wanted to know which companies offered such tests, the prices, and the number of SNPs tested. There is currently no central website comparing all the services that are on offer. I also wanted to have some idea of what I might expect from the test. This information was provided by 23andMe as I was able to set up a demo account to see how the test worked. The number of SNPs tested, whether I would have access to the raw data, and the quality of the company's analysis and website. The type and extent of the analysis. How many SNPs within the TS gene of interest were reported by this test? How dense was	Number of variants tested SNPs tested Number of SNPs tested Comparison in regards to significance with SNPs -not on chip -on other chips Cost of test Number of SNPs tested What to expect/demo version Number of SNPs tested Consumer access to raw data Quality of analysis Type of analysis Extent of analysis Number of SNPs tested within a specific gene Density of coverage along the chromosome adjacent to a

	the coverage along the chromosome adjacent to this gene? Were the genealogic reporting functions of the company adequate to follow inheritance of the "IBD" segment containing the gene? The latter was as important as the others.	specific gene Possibility of following the "IBD" segment containing a specific gene
	Would I have access to the raw data.	Consumer access to raw data
	cost, what the test would tell	Cost of test What information the test would provide
	Cost, privacy, ongoing updates, ability to find relatives.	Cost of test Updates to results
	how many traits and health conditions would be covered, what I'd be likely to learn of my genealogy	Number of health conditions covered
	accuracy and what information you could get and costs were main things Also checked years in business reputable top companies etc	Accuracy of tests Information provided Cost of test Years in business
	The turn times & the nature of the data produced.	Time taken Nature of data produced
	How the test is done (swab, gum, spit); what would I learn; privacy and security were of minor importance.	How to do the test Information you can learn from test Privacy issues Security issues
	Cost, results provided	Cost of test Results provided
	Price, conditions covered, whether interpretations will be updated, how data will be used (e.g., for research).	Cost of test Conditions covered Updates to interpretations How data will be used by company
	I wanted to know what genes for T2d were tested for.	Genes tested for a specific condition

	Price; expected results(i.e.- what would be revealed); size of company's database; help interpreting results to be expected.	Cost of test Results provided Size of company's database Whether there would be help interpreting results
	# of SNPs. Third party tools I can run like BGAs, Prometheus etc.	Number of SNPs tested
	what diseases/ traits were tested cost of test vs. information to be learned	Diseases tested Cost of test Information provided
	I wanted to know the specific application(s) for the results.	Specific applications of the results
	Accuracy. Description of what results are expected.	Accuracy of results Description of results
	Cost, method of providing sample, timescale for results, view of example results.	Cost of test Method of providing sample Time taken Example results
	I wanted to know if the initial test was thought to be reliable.	Reliability of test
	Number of SNPs. Links to authoritative medical sources for any apparent correlation between an allele and a disease/condition.	Number of SNPs tested Links to evidence for disease/gene associations
	Reliability, price, accuracy, which diseases and traits are presented in results.	Reliability of test Cost of test Accuracy of test Results for which diseases
	genotyping method genes tested algorithm for interpretation of results	Method – genotyping Genes tested Algorithm for interpretation
	cost, financial stability of testing company, reputation of testing company	Cost of test
	I wanted to know if it was reliable - I wrote to "Ask the geneticist" at their website.	Reliability of test

	They were lukewarm, but not negative. Seemed to be a good gamble.	
	That it couldn't be used to increase the cost of my private health insurance.	Situation in regards to health insurance
	I was mostly interested in know how serious the company is (23andme) and they are doing to ensure high quality on their test	Quality control procedures
	how to take the sample, what it would tell me & price	How to take sample What test would tell you Cost of test
	I checked the price and the number of SNPs tested. I specifically checked if genes related to specific traits (such as red hair) would be tested.	Cost Number of SNPs tested
	1) if the results were repeatable 2) if similar tests from different companies were comparable 3) the difference between SNP- and STR-based tests and how many probes each have 4) what technology is behind them (the testing chips) 5) what the business model behind the companies were and how they hoped to make money from them	Repeatability of results Chips used Business model of company
	# of genetic markers analyzed, # of conditions reported, whether data would be updated over time, whether I would have access to raw data, privacy policy	Number of genetic markers analysed Number of conditions reported Updates to results Consumer access to raw data Privacy policy
	How many SNPs would the test cover?	Number of SNPs tested
	Cost Confidentiality Value for money	Cost of tests Confidentiality of results
	price usefulness	Cost of test Usefulness of results

	I wanted to know from my results, which autosomal diseases would be identifiable in terms of my carrier or 'afflicted' status, and whether new results and/or research outcomes would be compatible with my genomic information from 23andMe.I also wondered whether 23andMe would facilitate interpretation of my results in regard to those issues. Wanted to know what parts of my DNA would be tested and what standards they would be compared to. How much of my genome was tested. accuracy procedure what kind of information would I get from the test I wanted to know the difference between a complete DNA sequence and a list of SNPs (which is what 23andme.com provided). Would my information be sold or otherwise exploited Cost, available genes that were being tested My primary incentive for purchasing the tests was genealogical in nature so I wanted to be sure I could share genealogical data without revealing medical info. I also inquired into the confidentiality and privacy controls for all the data.	What diseases are covered Updates to results Help with interpretation of updates What parts of DNA tested What is data compared to How much of genome tested Accuracy of test Procedure used Information you would get from test Difference between a completed DNA sequence and a list of SNPs Would information be sold or similar Cost of test Genes that are tested Confidentiality Privacy controls Consumer access to raw data

	access to results Intrepretation of results Security of results, accuracy Cost, method and sample of results interface (website) What it was actually doing - i.e. SNP chip What would be available - analysis + raw data What the test provides, types of information it gives. I was most worried about privacy. Information about disease and ancestry was about equal value. I was curious how the DNA would be collected. I had to drool into a plastic container, kinda gross. As I said, I was curious. I was curious about the process, the results, the business model, the whole thing. - How much can be learned from snips - Will I have access to my raw data so I can have 3rd party analysis. (answers: A bit, not as much as I'd hoped and Yes. How complete is the genome coverage (i.e., how many SNPs does their microarray have) Privacy concerns What the results meant; how long until the results were available. size of genetic coverage (i.e. it has to include more than	Interpretation of results Security issues Accuracy of test Cost of tests Methods used Sample of user interface SNP chip used Consumer access to raw data Consumer access to analysis Information provided by the test Privacy issues How the sample is taken Testing process Results What can be learned from SNPs Consumer access to raw data How much of genome is tested Number of SNPs tested Privacy issues Meaning of results Time taken What genetic material (i.e. chromosomes, mitochondria) is tested

	mitochondrial and sex chromosomes)	
	actual test report	Example report
	What exactly was involved in getting the data to them, how long it would take to get results, the accuracy of the tests involved	Sample process Time taken Accuracy of tests
	Privacy policy. Price.	Privacy policy Cost of test
	How long it would take to process, how the tests determine the information they provide.	Time taken Method of analysis
	what conditions were tested for; the estimated accuracy of the results; how the data (and genetic samples) were maintained with respect to privacy.	What conditions are tested for Accuracy of results Privacy of data Privacy of sample
	How complete the test would be (in other words, how much of my genetic profile would be indexed), what kinds of information would be provided, what kinds of analysis of the data would be provided by the testing company.	How much of genome sequenced Kinds of information provided Analysis of data
	cost, the system / method of how they going to display the results, who owns the company, the process how they collect my sample	Cost of tests How results are displayed Who owns the company Sample process
	What the process would be for me, what they did with my genetic material, what information would be made public, how they found results	Test process Fate of genetic material What information made public Method of analysis
	security of information and privacy. 23andMe has a pretty good repuation.	Security of information Privacy issues

	how detailed they get how accurate are they	Detail of results Accuracy of test
	their approach to presenting the data, their approach to confidentiality, how many markers were typed	How data is presented Confidentiality issues
	Type of equipment. Process. Reliability of results. Reputation of vendor/lab. Privacy/security of data.	Equipment used Testing process Reliability of results Privacy issues Security issues
	How it worked.	Methods
	Privacy, accuracy, level of detail.	Privacy issues Accuracy of test Level of detail
	i wanted to know if it is a valuable resource.	Usefulness of results
	How much, how reputable, how many sections they were testing, how private.	Cost of test How much of genome is tested Privacy issues
	How many SNPs were covered, what sort of ancestry analysis was available	Number of SNPs tested
	If it was safe, secure (privacy), accurate	Safety of tests Security issues Privacy issues Accuracy of test
	How comprehensive the test was, how other users felt about the test, what the price was.	Comprehensiveness of test User reviews Cost of test
	what's required (saliva sample), price, what it can tell me	Sample process Cost of test What it can tell you
	what they're testing for how they're protecting my information (or trying to :)) cost	What the test covers Security issues Cost of test
	Mostly about the	Confidentiality issues Consumers access to raw data

	trustworthiness of the company: Confidentiality; whether raw data was allowed to be downloaded/extracted by the user; whether there is a hidden agenda or unexpected ways the data might be used; how does the company expect to make money off the data if they are offering the test so cheaply cost, what diseases it covers How long it takes What is the risk of contamination Which conditions they test for What my data is used for and how it's used. How the genetic analysis is performed technically and its scientific background.	How the company uses data Cost of the test What diseases are tested Time taken Risk of contamination What conditions are tested for How company uses the data Method of analysis Scientific background of analysis
[Consumers' questionnaire] 14. Please describe briefly what you did when looking for information about genetic tests, including what sources you used, what information you were looking for and how successful you were.	Primarily online DNA forums and mailing lists--i.e., reports from other customers I considered the knowledge of "experts" and who was associated with 23andMe as well as who used the service mainly looking for what was covered and the price, and was fairly successful using the internet Google search and compared prices and what diseases were screened. Asked around to network of biologist colleagues, read blog	Customer reviews Who is associated with the company What the test covers Cost of test Cost of test What diseases are tested Cost of test What the test covers

	and twitter accounts, other company web sites. Looking for cost, data coverage. Found it.	
	Feedback & reports by others about their experience. Blog posts & reviews by scientists & other trusted sources.	User reviews
	Researched science and technology blogs regarding offering firms reliability, price, and usefulness of test results.	Reliability of test Cost of test Usefulness of results
	look at the genes & health conditions, the bibliography, and, very important, sample reports	Genes looked at Conditions looked at Bibliography Sample reports
	colleagues, journals, internet, work, existing data, prices	Cost of test
	looked at wikipedia talked to genetic counsellors looked for guidance on how to interpret results	Results interpretation
	Read papers on the subject, looked up details on the chip from Illumina	Chip details
	The 23andMe test was an absolute breakthrough. NO ONE ELSE DID IT. Once I found that I could use it for my purpose (mentioned above -it covered the region of interest densely enough), the only issue was price. It took some effort to find a discount that would make the project affordable, since we needed to test 7+ individuals	Cost of test
	cost and access to raw data for 3rd party analysis	Cost of test Consumer access to raw data
	I read user's ratings of the site and the nature of the information that would be produced.	User reviews Nature of information provided

	I went to the scholarly literature on the genetics of type 2 diabetes.	Genetics of a specific disease
	I looked for testimonials from others and the responses varied quite a bit. Often those "not very satisfied" were found to be ignorant about the result meaning.	User reviews
	looked at price, ancestry information. very satisfied.	Cost of test
	I made google searches on the technologies they used, the algorithms they used, and on the publishing records of the scientists they hired.	Technology used Algorithms used Publishing records of the scientists
	Before I purchased a test for myself and one of my sisters (on special) I looked at whether the results could be used for analysis by other sites/organisations to provide information or analyses not conducted or provided by 23andMe. I also checked whether any of the results would be immediately useful when checked against recent/current scientific and medical journal articles and databases.	Consumer access to raw data Immediate usefulness of results
	Type pf tests high number of datum	Type of test
	Looked for accuracy of testing, found news articles that made me believe the testing was pretty accurate	Accuracy of tests
	method, size of information I can get	Methods – general Amount of information produced
	read papers on genetics & diseases, looked at the	Information on results Cost of test

	websites of different direct-to-customer-genetic-services, looked for best result/price ratio Googled information for legitimacy, reliability, and long term commitment to customers. Also ability to answer any questions after receiving results. It was pretty haphazard, to be honest. Having read glowing endorsements from users on blogs (that I hadn't specifically sought out), all I wanted from the specific company I chose was very basic information, such as the price. This was a process of research on the internet, joining email list and discussing the subject to understand what tests do what and realise that first I needed to decide what I wanted to find out before ordering the first test.	Reliability of tests Will there be a long term commitment Answer consumer questions Cost of test What the tests do
[Potential Consumers questionnaire]6. What information about genetic tests do you want to know before purchasing a test? i.e. information about the test itself, NOT your reason for purchasing the test. (Please list as many answers as you can)	I would like to know about every data they can collect from my genome and wich techniques and basic genetics knowledge support their analysis. Besides that I would like to know how secure they are going to be about my genetic information because I don't want to share it with people that I don't know. Privacy (can my information be	What the test covers Methods of analysis Evidence for analysis Security issues Privacy issues

	accessed by insurance, the government, or other organizations?)	– insurance -government -other third parties
	how the information is stored and kept secure	Storage of data Security of data
	What technique they use, how long it takes to complete the process	Methods Time taken
	Breadth of data collection and quality and objectivity of statistical analyses.	How much the test covers Quality of analysis Objectivity of analysis
	accuracy	Accuracy of test
	How many people have taken it, what sorts of things I can compare my results to, whether they help you analyze it or not	Number of users What results can be compared to Help in interpretation
	what is the turnaround time what is the error/accuracy rate	Time taken Error rate Accuracy of tests
	How does it work? How long will it take to receive results? What resources does the company for understanding the results?	How test works Time taken What is analysis based on
	Total cost # of markers Platform Done in certified lab Data return	Cost of test Testing platform Lab certification How data is returned to consumer
	Cost, time until results, accuracy	Cost of test Time taken Accuracy of results
	What will the result package look like. How are the results presented. How detailed is it. How accurate are the results. Are some parts more accurate than others.	How will results be presented Sample results How detailed are results Accuracy of results Any variability in accuracy
	How comprehensive it is (# of	Comprehensiveness of test

	SNPs)	Number of SNPs
	which genes are included or not, certainty of analysis etc	Which genes are covered Confidence in analysis
	How accurate the gene sequence is, how detailed a look do they perform, what method is used to determine the gene sequence and what are the limitations of that method	Accuracy of sequencing Detail of sequencing Method of sequencing Limitations of sequencing method
	what they can find out - and what not	Information that can be found out Information that can't be found out
	What it can give me, what I will know	Information provided What can be learnt from test
	1. I want to be able to download the raw data. 2. Want to know how the keep the sample I send to them. 3. If they upgrade their service later, how that would effect me.	Consumer access to raw data Storage of sample Upgrade information
	1. Is the data mine? 2. Will the company handle the data in a private manner? (they won't share it without my permission) 3. Will the company provide explanation of the data to the level that an educated layman would understand? 4. How reliable is the test?	Who owns the data Privacy issues Third party access to data – permission Is appropriate explanation of data provided Reliability of test
	How accurate are the tests, single SNPs most of the time don't yield that accurate results and only indicators.	Accuracy of test
	What techniques they use, whether the lab meets FDA etc approval (or whatever the relevant body is that regulates that suff!).	Methods used Lab certification
	How it works What conditions they can test for What support	How the test works Conditions covered

	they offer before and afterwards, both for me and my family The cost of the test Info on what treatments they have	Support -before -afterwards -individually -family Cost of the test
	I would be most interested in how reliable the results might be.	Reliability of results
	Security of information.	Security of data
	How are the statistics derived? What is the external validity of these tests? What is the probability the "probabilities of getting a disease" are actually correct?	Method of analysis Validity of tests Accuracy of predictions
	Assured accuracy	Accuracy of tests
	If the lab is proffesionally certified, if their science is sound and not quackery, if they have any negative reviews and/or fraud/scam claims. What sort of testing they are offering, how many SNP's they are going to check, what sort of credentials their lab staff has.	Lab certification/credentials Validity of science Type of testing Number of SNPs
	I really need to know more about what I might be able to learn beyond what I already know and whether there would be any value to that additional learning. I haven't done any serious research on it yet. I haven't reviewed any websites on the matter yet.	What you can learn from the test
	Security and privacy. How extensive the tests are.	Security issues Privacy issues How extensive the tests are
	How it works (ie, what kind of genetic material to be collected, how its processed, how results are presented) and data security (who will have	Sample process How sample is analysed How results are presented Security issues Third party access to data

	access to my records once this information is collected?).	
	how long the data is stored, and how easily it is to be identified with me personally, and whether there is potential for the information to be shared.	Time data is stored for How easily are consumers identifiable from data Can data be shared
	How confidential are the results.	Confidentiality of results
	Who's doing the testing. What inherited conditions are tested. What are the disease risk percentages.	What laboratory What conditions tested
	What the test can tell me about specific risk factors, level of detail, which results are actionable	What the test can say about specific risk factors Detail of test Which results are actionable
	I would like to know how I can use the information afterwards.. How will the results be illustrated to me. How easy is it to access. How long does it take. Do you store my information for statistical use. Can I acces it form, lets say an iPad or Smartphone? - Do you have an app? Can anyone finde me using that data. Can I remove my data from your ownership. etc. (Hope that was usefull)	How can the information be used How are results presented How easy to access results Time taken Is information stored for statistical use What platforms can you access data from Is there an app Can consumer be traced from data Can data be removed from company
	Reliability. Accuracy. Confidentiality.	Reliability of test Accuracy of test Confidentiality issues
	Accuracy of data; reliability; value (i.e. will the test give me results that are meaningful... not just the obvious traits and statistical likelihood of disease but specific susceptibility)	Accuracy of data Reliability of data Usefulness of data Specific susceptibility
	I want to see outside verification of the test's	Outside verification of accuracy Breadth of genetic information

	accuracy, that it covers a wide range of different genetic information, that the price is comparable to other services, and that the results are easy to understand.	covered Cost of test Are results easy to understand
	reliability of the test	Reliability of test
	Whether the raw data can be downloaded, what the error rate is, how comprehensive is the testing,	Consumer access to raw data Error rate Comprehensiveness of test
	I'd like to be able to examine a sample test to see exactly what I'd be learning, how the information is presented, and how accurate they can guarantee the information to be.	Sample results What can be learned from test How information is presented Accuracy of information
	Accuracy; price; format of results; will any 'analysis' be provided by company doing the WGS. (I don't really want to pay extra for any.)	Accuracy of test Format of results Will analysis be provided by the company
	What does the test tell me, and how does it affect my insurance (in the US if you get a genetic test it can affect your ability to gain insurance due to "pre-existing conditions")	What does the test tell you Does it affect insurance
	accuracy reliability privacy	Accuracy of test Reliability of test Privacy issues
	Confidence intervals on results; what type of results I will be receiving i.e. what will it be testing against. Paired preventative measures with results that are found to have high likelihood.	Confidence intervals of results What the results are for Preventative measures
	Would the company give potential employers or health insurance companies my results? I know that	Access to results by third parties -insurance -potential employers

	theoretically I cannot be denied a job or health insurance because of the results, but I don't want them to have access to my results at all. I have privacy concerns.	Privacy issues
	How diagnostic they will be - especially since I have kids and want to know if there are specific risk factors to look into	How diagnostic are the tests
	how information may be used/shared	How information may be used How information may be shared
[Potential Consumers questionnaire] 9. Please describe briefly what you did when looking for information about genetic tests, including what sources you used, what information you were looking for and how successful you were.	I looked at price mainly, data is data, and the quality of data from the DTC cos are roughly equivalent - many use the same technology.	Cost of test
	Platform, wanted to know if I could get the raw data. Just asked people who had already done it.	Platform used Consumer access to raw data
	I searched blog posts, examined the Wikipedia articles regarding the methodologies, and spoke with my wife regarding it, who laughed that I would think of wasting money that way.	Methods
	Started to follow Navigenics on twitter. i went on their site curious about how they did their tests and read about them.	Method of testing
	Confirm the availability of my data after I take the test. (Can I download my SNPs?) I've talked with friends about their experience.	Consumer access to raw data
	I mainly Googled around the terms, looking at journal articles and wikipedia pages to educate myself on the material described by these companies. I was looking to understand the	Reliability of tests Validity of tests

	tests and make a judgement on their reliability and validity. I was somewhat successful, but not satisfied enough to make a decision whether to buy or not buy.	
	I look for: I heard that same drug work differently in different people. I want to make sure that I got a satisfied answer but its never was.	Information about pharmacogenomics
	Looked for company who had the largest data base and had the most support for any questions	Size of databse Question support
	I read about 23 in Wired, checked out the website and became very interested. I just wanted to know what types of diseases they check against, confidence intervals, etc.	Conditions covered Confidence intervals for results

Items Generated from Professional Guidelines and Recommendations

Organisation	Website	Recommendations	Distilled
The American Society of Human Genetics	http://www.google.co.uk/url?sa=t&rct=j&q=ashg%20statement*%20on%20direct-to-consumer%20genetic%20testing%20in%20the%20united%20states&source=web&cd=1&ved=0CFAQFjAA&url=http%3A%2F%2Fwww.ashg.org%2Fpdf%2Fdtc_statement.pdf&ei=xxC6T-arMLST0QW159nnBw&usg=AFQjCNE6ko3KgE_vNQc__D5-zF92NH3xUQ	To promote transparency and to permit providers and consumers to make informed decisions about DTC genetic testing, companies must provide all relevant information about offered tests in a readily accessible and understandable manner. a. Companies offering DTC genetic testing should disclose the sensitivity, specificity, and predictive value of the test, and the populations for which this information is known, in a readily understandable and accessible fashion. b. Companies offering DTC testing should disclose the strength of scientific evidence on which any claims of benefit are based, as well as any limitations to the claimed benefits. For example, if a disease or condition may be caused by many factors, including the presence of a particular genetic variant, the company should disclose that other factors may cause the condition and that absence of the variant does not mean the patient is not at risk for the disease. c. Companies offering DTC testing should clearly disclose all risks associated with testing, including psychological risks and risks to family members. d. Companies offering DTC testing should disclose the CLIA certification status of the laboratory performing the genetic testing. e. Companies offering DTC testing should maintain the privacy of all genetic information and disclose their privacy policies, including whether they comply with HIPAA. f. Companies offering DTC testing and making lifestyle, nutritional, pharmacologic, or other treatment	Sensitivity of the test Specificity of the test Predictive value of the test Populations for which this information is known Evidence for benefits Limitations in regards to claimed benefits Risks – general Risks – psychological Risks – family members CLIA certification of laboratory Privacy policy – general Privacy policy – HIPAA Evidence for recommended interventions Evidence against recommended interventions

		recommendations on the basis of the results of those tests should disclose the clinical evidence for and against the efficacy of such interventions, with respect to those specific recommendations and indications.	
The American College of Clinical Pharmacology	http://jcp.sagepub.com/content/49/8/886.long	What is the population at risk for the disease that is the focus of the advertisement, and what percentage of individuals with that disease actually has a strong genetic component to the disease's expression? Recognize the scientific limitations of each test. Realize that many companies that sell DTC genetic testing services do not provide interpretation of test results. Pre- and posttest counseling and result interpretation must be sought by the consumer.	Population at risk for disease Percentage with disease whose disease expression had a strong genetic component Limitations of the test Availability of counselling pre-test Availability of counselling post-test Statement of who interprets results (e.g. consumer or counsellor)
The European society of human genetics	http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3002858/?tool=pubmed	Among other issues, the advertisement should be accurate and not misleading, claims should be transparent and supported by current evidence, and complete and accurate information about the test limitations, risks and benefits should be provided. Key concerns are the provision of sufficient information about the purpose and appropriateness of testing, its possibilities and limitations, as well as the clinical significance of testing. The labelling information on genetic tests must be true, accurate, accessible, complete and comprehensible.. Privacy and confidentiality of the results, as well as possible consequences related to their disclosure to third parties, such as insurance companies and employers, should be discussed, when appropriate, as well as the property of the biological material and its fate after the results are confirmed. [Companies should] inform [consumers] about their	Limitations of the test Risks of the test Benefits of the test Purpose of testing Appropriateness of testing Possibilities of testing Clinical Significance of testing Privacy and Confidentiality Risks of disclosure of information -general Risks of disclosure of information – web community Property of biological material Fate of biological material – general Fate of biological material – if company sold or bankrupt Security procedures

		security procedures, explain what will happen to the sample and the data when the testing process is concluded, and have a clearly laid-out plan as to what will happen to the samples and data should the company be sold or go bankrupt. Companies inviting their customers to share their genetic information via a web community or forum should inform people about the potential risks for disclosure of this type of sensitive information. If samples or data are to be used in any research, this should be clear to consumers, and a separate and unambiguous consent procedure should take place. Informed consent documents for participation in research should disclose the procedures for storing and disposal of samples and genetic information, the time period and conditions for storing them, inform participants of the identity of any third parties who may be granted access to data or samples, and include also information on the fact that the research may lead to commercialization and patents, on any customers' rights to commercial benefits and on the property of biological samples and data.	Fate of data – general Fate of data – if company sold or bankrupt If samples or data used in research - Stated this happens - Procedure for storage and disposal - Time period and conditions of storage - Identity of third parties with access - Possible lead to commercialization and patents - Customers' rights to commercial benefits - Property of samples and data
International Society of nurses in genetics	http://www.isong.org/ISONG_PS_direct_consumer_marketing_genetic_tests.php	Therefore, it is the position of the ISONG that utilization of DTC genetic testing be undertaken once the consumer has considered, independently, or with the help of a professional, the following; 1. The privacy mechanisms in place to ensure confidentiality of genetic information; 2. The purpose of the test and how the results will be used; 3. The clinical value of the test, or if and how the results can inform choices with regard to health care, behaviors, and lifestyle;	Privacy Issues Purpose of test How results will be used Clinical value of test If results can help inform health behaviour choices How results can help inform health behaviour choices Concerns over testing of children Scientific usefulness of tests Laboratory issues Accuracy of interpretation of

		4. The additional concerns posed in the testing of minors; 5. The ability of the test results to provide scientifically based information relevant to the reason the test was requested; 6. The reputation of the company offering the testing in light of the fact that, depending on geographic location and the specific test, companies may be functioning with little, if any, regulatory oversight; 7. The quality/reputation of the laboratory performing the testing to assure the accuracy of the test and whether one can ascertain this information; 8. The accuracy and adequacy of the interpretation of the results; 9. The fate of the genetic material (destroyed, stored or used for research) after the test is complete. The use of genetic material for research purposes should be transparent and permission obtained; and 10. The potential benefits derived from genetic counseling prior to and after genetic testing to determine the appropriateness of the test and to explore the meaning of the results for the individual and the family.	results Adequacy of interpretation of results Fate of genetic material If genetic material is used for research – should be stated - Permission should be gained Potential benefits from genetic counselling – pretest - Post-test
American College of Medical Genetics	http://www.google.co.uk/url?sa=t&rct=j&q=statement%20direct%20to%20consumer%20genetic%20test&source=web&cd=12&ved=0CIUBEBYwATgK&url=http%3A%2F%2Fwww.acmg.net%2FStaticContent%2F	**The consumer should be fully informed regarding what the test can and cannot say about his or her health.** Many DTC genetic tests do not give a definitive answer as to whether an individual will develop a given condition, but provide only a risk or probability of developing a disease. The interpretation of such results is	What the test can say about health What the test can't say about health Appropriate communication of results, indicating their highly

	StaticPages%2FDTC_Statement.pdf&ei=oyy6T4nfIunD0QWO9ejCDg&usg=AFQjCNG52cDLmrcZEleQVifuzbUWedaYZg	often highly nuanced and such information needs to be communicated to the consumer in the appropriate context and in an understandable fashion that is linguistically and culturally appropriate. • **The scientific evidence on which a test is based should be clearly stated.** DTC genetic test providers should provide easy-to-understand information with primary references documenting the scientific data on which a specific test is based. • **The clinical testing laboratory must be accredited by CLIA, the State and/or other applicable accrediting agencies.** The accreditation process ensures that laboratories adhere to strict standards and guidelines for clinical testing. Test result reports to consumers should indicate the specifics of the lab's accreditation. • **Privacy concerns must be addressed.** Prior to testing, the consumer should be informed regarding who will have access to test results, what security is in place to protect these results, what will happen to the DNA sample once testing is complete and how to access a complaint procedure to report breaches of privacy. Also, the issues of possible employment and insurance discrimination and the potential impact on other family members should be discussed prior to obtaining genetic testing.	nuanced nature Scientific evidence on which specific tests are based Primary references for scientific evidence on which specific tests are based Laboratory accreditation Privacy issues – who will have access to test results - Security in place to protect results - Fate of DNA sample - How to access complaints procedure about breaches of privacy Risks – employment discrimination - Insurance discrimination - Family members
The American Congress of Obstetricians and	http://www.acog.org/Resources_And_Publications/Committee_Opinions/Committee_on_Genetics/Direc	However, patients are not made aware that failure to indicate results of genetic testing in life insurance or disability applications could be considered fraud. In	Legal position of declaring results for insurance Legal position of declaring results

Gynaecologists	t-to-Consumer_Marketing_of_Genetic_Testing	addition, many laboratories have not indicated their policies on what is done with the DNA sample after analysis. To ensure privacy, DNA samples should be destroyed after the requested test is performed. Those overseeing procedures for testing should continue to work to address patient privacy concerns. Appropriate pretest and posttest counseling should be provided, including a discussion of the risks, benefits, and limitations of the testing.	for disability applications Fate of DNA sample Privacy issues Risks of testing Benefits of testing Limitations of testing
The American Society of Clinical Oncology	http://jco.ascopubs.org/content/28/5/893.short	In the absence of genetic counseling by their health care providers, it is necessary for individuals seeking DTC testing to proactively obtain information they need to make informed decisions. The basic elements of consent identified by ASCO can serve as a framework for gathering this information. Awareness of laboratory privacy policies and practices related to data security, laboratory compliance with applicable licensing requirements, the availability and cost of genetic counseling, and the possible use of DNA testing samples in future company research may be relevant to an individual's decision to pursue genetic or genomic testing. Companies offering DTC tests should make this information clearly and easily available to the public, preferably as part of a consent form that must be acknowledged by the individual undergoing testing before testing is completed. Laboratories intending to conduct research using DNA samples submitted for testing should obtain consent to use these samples. The consent form should explain whether and how samples will be identified, stored, and destroyed, and whether genetic risks found through future research will be reported back to individuals who allow their samples to be used. Testing should not be contingent on allowing DNA samples to be	Privacy Policy Data security Laboratory licensing Availability of genetic counselling Cost of genetic counselling If DNA sample may be used in research - it should be stated. - Whether samples will be identified - If samples are identified then how - Storage of samples - Whether samples will be destroyed - Communication of genetic risks Availability of cancer genetic risk assessment Potential risks – life insurance - Disability insurance - Long-term care insurance How data can be shared with outside parties

		used in future research. Testing laboratories should make information about data privacy, data security, laboratory licensure, the availability of genetic counseling or cancer genetic risk assessment, and any potential for future use of DNA samples submitted for testing clearly and easily available to the public. However, it is important for patients to be aware that, at this time, there are no special protections against the use of genetic information to inform the provision of life insurance, disability insurance, or long-term care insurance. Individuals considering DTC testing should become familiar with the terms of company policies related to privacy and data security, including how genetic information can be shared with outside parties or become part of their medical records. ASCO recommends that individuals considering genetic testing become familiar with company policies related to privacy and data security. Laboratories providing testing should develop written privacy policies that are easily accessible to individuals considering testing. Any claims about the privacy of DTC testing should be truthful and nonmisleading.	If data can become part of medical records
Austrian Bioethics Commission	http://www.bka.gv.at/site/4070/default.aspx	§ 69 (1) GTG stipulates genetic analy-ses of the aforementioned types 2, 3, or 4, including any analysis carried out as part of a prenatal examination , may only be carried out with the written consent of the person to be examined. Furthermore, the patient must have been given prior medical advice regarding the nature, scope and informa-tive value of the test by a specialist physician trained in human genetics/medical genetics, or a physician specialising in the relevant medical field. The person to be tested must have freely consented to the genetic analysis	Nature of the test Scope of the test Informative value of the test Limits to informative value Possible risks Possible dangers Data protection issues Precise purpose of the test What the test entails

		on the basis of the full information given to him or her. Genetic counselling after a genetic test must include a factual and comprehensive discussion of all test results and medical facts and as well as of the possible medi-cal, social and psychological consequences. In the case of a disposition to a hereditary illness with grave physical, psychological and social effects, tested persons must be advised in writing that it may be advisable to seek additional non-medical counselling by a psychologist, psychotherapist or social worker. Information can also be provided about other counselling facilities and self-help groups. Pre- and post-test genetic counselling must be non-directive. At the beginning of the counselling session, the individual seeking advice must be told that they may at any time – even after they have consented to the genetic analysis, or after the counsel-ing – announce that they do not wish to be nformed of the results of the analysis and the resulting consequences. Pre- and post-test genetic counselling must conclude with a personal letter sent to the advice seeking individual summing up the most important areas covered by the counselling session in an accessible manner. The law also requires that the patient's relatives, or the test results and other data pertaining to them, are dealt with in a diligent and careful manner. The essen-tial prerequisites for this are that the genetic information offered is correct and that indi-viduals interested in taking a genetic test are adequately informed about the nature of the tests, the limits to their informative value, possible risks and dangers, and data pro-tection issues. Given the complexity of the material, it is doubtful that this is possible without appropriate specialist counselling. However if a person decides to underg such Internet-based	

		genetic testing fo multi-factorial conditions, they shoul obtain information about the precise purpose of the test, and what it entails.	
Belgian Advisory Committee on Bioethics	http://www.google.co.uk/url?sa=t&rct=j&q=opinion%20no.%2032%20of%205%20july%202004%20on%20the%20free%20availability%20of%20genetic%20tests.%20belgian%20advisory%20committee%20on%20bioethics&source=web&cd=1&ved=0CEkQFjAA&url=http%3A%2F%2Fwww.health.belgium.be%2Ffilestore%2F13084478%2FOpinion%252032%2520web_0_13084478_en.pdf&ei=7Mq6T_-WL-au0QWy-sT5Bw&usg=AFQjCNEizxolY3Em0HkWlAnhcLBybtmroQ	A second group of members thinks that a blanket ban on free access is not desirable and at the same time is not feasible. They feel that the tests should be provided with an information leaflet containing at least the following details: - the aim of the test; - the limitation in the interpretation of the results; - information in which it is recommended that people receive genetic counselling; - the contact details of the eight recognised genetic centres in Belgium. This instruction leaflet should moreover recall: - that the results of the test can be important for the relatives of the person requesting the test; - that the right to know as well as the right not to know should be respected vis-à-vis these people, too. This leaflet should be written in easily understandable language.	Aim of the test Limitations of the interpretation of results Recommendation for genetic counselling Contact details of genetic centres State that the results can be important for relatives That relatives have a right to know That relatives have a right not to know
National Council of Ethics for the Life Sciences (Portugal)	http://www.google.co.uk/url?sa=t&rct=j&q=national%20council%20of%20ethics%20for%20the%20life%20sciences%20portugal%20genetic%20test&source=web&cd=6&ved=0CGAQFjAF&url=http%3A%2F%2Fwww.ethical-fp7.eu%2Findex.php%3Foption%3	...quality, transparency (including identification of the laboratory where the test was actually performed, genes and mutations tested, methods used, possibilities and limitations), as well as the test claims and the expectations they create, are aspects of eminent ethical nature The prior information needed to take a decision to perform a genetic test should be made	Identification of laboratory Genes tested Mutations tested Methods used Possibilities of tests Limitations of tests Sensibility of test Specificity of test

	Dcom_docman%26task%3Ddoc_download%26gid%3D50%26Itemid%3D78&ei=o9O6T7vSNM-l8gOtor3CCg&usg=AFQjCNF_fLAFO_-jdY1nrfNqat9wVOg45Q	available in a clear, accessible form, and should include the sensibility, specificity and predictive value of the test, the scientific evidence available for that particular population and the possible implications of its results for the persons tested and their family members.	Predictive value of test Scientific evidence available for population Possible implications for consumer Possible implications for family members
Nuffield Council on Bioethics	http://www.google.co.uk/url?sa=t&rct=j&q=nuffield%20council%20on%20bioethics%20personalized%20healthcare%20%E2%80%93%20chapter%209.&source=web&cd=1&ved=0CGMQFjAA&url=http%3A%2F%2Fwww.nuffieldbioethics.org%2Fsites%2Fdefault%2Ffiles%2FMedical%2520profiling%2520and%2520online%2520medicine%2520-%2520the%2520ethics%2520of%2520'personalised%2520healthcare'%2520in%2520a%2520consumer%2520age%2520(Web%2520version%2520-%2520reduced).pdf&ei=7tS6T_KpMIbS8gP7ysiXCg&usg=AFQjCNGddQGvNFQHpOefJTyK8HTh7SzeqA	...government health service websites should provide public information about genetic profiling services and companies should indicate to consumers where to find this information [in regards to screening] evidence-based information, explaining the consequences of testing, investigation and treatment, should be made available to potential participants to assist them in making an informed choice; We recommend that appropriate publicly-funded health service websites should include general information for the public about direct-to-consumer genetic profiling services provided by commercial companies. This information should include reference to: ■ potential risks and benefits; ■ any difficulties with establishing clinical validity; ■ the possibility of finding out about conditions for which treatment is not available; ■ the special case of children (see also recommendation in Paragraph 9.54); and whether it could be necessary for consumers to inform life, mortgage or travel insurance companies of the results of any tests, either at the time or in the future. We further recommend that governments should require details about where to find this	Link to government health service websites with information about genetic testing Consequences of testing Consequences of investigation Consequences of treatment Potential risks Potential benefits Any difficulties with establishing clinical validity Possibility of finding out about conditions for which treatment is not available Issues to do with testing children Whether it could be necessary to inform companies of the results (currently or in future) – life insurance - Mortage insurance - Travel insurance Operator of services Location of operator Evidence for interpretations of results Limitations of the test Probabilistic nature of tests

		information to be included in the advertising and information provided by companies selling genetic profiling services in their countries (see also our recommendation in Paragraph 9.51). We recommend that all companies that provide genetic analysis for susceptibility to common multifactorial diseases should make the following information prominently available in lay language for the consumer before they buy: ■ the operator of the services; ■ the location in which the operator is based; ■ the evidence on which interpretations of the test results are based; ■ the tests' limitations, including the fact that they are probabilistic and based on current research results which may change; ■ that the test results may require interpretation by a qualified medical practitioner or genetic counsellor; ■ the possibility of finding serious health problems and revealing family genetic relationships; ■ the nature of the risk being communicated to the consumer, i.e. absolute or relative risk; ■ advice about whether it might be necessary for consumers to declare any results they receive as a result of genetic tests to their life, mortgage or travel insurance companies; ■ which other third parties, if any, have access to the information/data;	Based on current research which may change Tests may require interpretation by GP or genetic counsellor Possibility of finding serious health problems Possibility of revealing family genetic relationships Nature of the risk (either absolute or relative) If third parties have access to data, which Results should not be used alone for medical decision making Tests that do not meet clinical validity requirements should not be carried out in children Data security arrangements – general Data security arrangements – changes to administration Funding arrangements Advertising arrangements Fate of data if company goes into administration or changes hands

		■ that the results should not be used alone for medical decision making given their limited clinical validity; ■ that tests that do not meet the requirement of clinical validity should not be carried out for children (see recommendation in Paragraph 9.54); ■ arrangements for data security (including in case of any changes to the administration of the company); ■ funding and advertising arrangements; and ■ where to find independent information about this type of service on public healthcare service websites (see our recommendation in Paragraph 9.49). We further recommend that all companies selling direct-to-consumer genetic tests follow the Common Framework of Principles intended for international use by genetic test providers developed by the Human Genetics Commission and approved by the Department of Health in England. Genetic profiling companies should provide details about what would happen to personal genetic data and interpretations should the company go into administration or change hands. This information should be made available to consumers before they buy	
Human Genetics Commission	http://www.hgc.gov.uk/Client/document.asp?DocId=280&CAtegoryId=10	**Marketing and advertising2.1** Where relevant, the test provider should comply with any legislation or voluntary codes for advertising of medical tests, including genetic tests or other clinical services and they should also comply with more general guidance (including legal guidance)	Characteristics of the tests Limitations of the tests Not overstate utility Claims about clinical validity should be supported with an

		covering consumer advertising.**2.2** Promotional and technical claims for genetic tests should accurately describe both the characteristics and the limitations of the tests offered, and the test provider should not overstate the utility of a genetic test.**2.3** Where a claim is made about the clinical validity of a genetic test, the claim should be supported by relevant evidence published in peer reviewed scientific literature and the test provider should give standard references to this literature.**2.4** The test provider should be aware of the risk of bias when quoting evidence and ensure that evidence is presented transparently with reference to the criteria used to include and/or exclude published literature when this is cited as evidence of the applicability or effectiveness of the test.**2.5** Information about tests which are available only in the context of a consultation with a health professional or are only provided to consumers with both individualised pre- and post-test counselling should make it clear that tests are available only in that context.**3. Regulatory Information3.1** The test provider should make available the evidence of the association between a genetic marker and a disease, condition or trait for the genetic tests that they supply. Ideally, the associations should be validated at genome wide significance level in more than one large case control study and in a cohort of the ethnic/geographic background relevant to the client. The associations should be published in peer-reviewed scientific journals, they should be undertaken in line with the recommendations made in the STREGA statement*, and the provider should supply standard references for these publications. **Information for prospective consumers 4.1** The test provider should supply easily understood, accurate, appropriate and adequate information, which is also	appropriate reference Evidence should be presented transparently If evidence is presented there should be reference to the criteria used to include and/or exclude published literature If only available through healthcare professional or after counselling should be made clear Evidence of the association between a genetic marker and a disease, condition or trait General information about genetics Role of genes in health and disease Role of genes in conditioning phenotypes Technologies applied to generate the knowledge Relative roles in determining health and disease of - genetics - environmental factors - lifestyle choices - and other factors Relative roles in determining phenotype of - genetics - environmental factors - lifestyle choices - and other factors

		available in accessible formats, to consumers before obtaining consent for a genetic test. The following should be provided: L Lgeneral information about genetics to enable a consumer to understand the scientific basis of genetic testing, the role of genes in health and disease, and conditioning phenotypes, and the technologies applied to generate the knowledge L La clear explanation of the relative roles of genetics, environmental factors, lifestyle choices and other factors in determining health, disease and phenotype L Lspecific information about genetic tests offered L Linformation about counselling offered in connection with the test including whether counselling is included in the cost of the test and for what costs the consumer will be liable if they withdraw following pre-test counselling L Linformation about the presentation of results in statistical form, such as relative and absolute risk assessments or likelihood of inclusion/exclusion as a genetic relative, so that an individual can understand test results that are provided L Linformation about measures taken by the test provider and laboratories to ensure the confidentiality of personal records and security of biological samples L Linformation about the maximum period of storage of the biological sample and personal records, and procedures for storage, transfer and disposal of biological samples and personal records L Linformation about whether biological samples may be used for any secondary purposes, such as additional research purposes, and about or whether personal genetic information may be passed on to third parties and, if so under what conditions and to whom L Linformation about procedures for handling and resolving consumer complaints L Linformation about the manner in which the test results will be provided and, if applicable to the genetic test, information about the	Specific information about tests offered Whether counselling is included If so, will consumers be liable if they withdraw following pre-test counselling Information about the presentation of the results -relative risk assessments -absolute risk assessments Confidentiality measures Security of samples methods Maximum period of sample storage Maximum period of records storage Procedures for samples -storage -transfer -disposal Procedure for records -storage -transfer -disposal If samples may be used for research then should be - stated If information may be passed onto third parties should be -stated -to whom - under what conditions Complaints procedures

		requirement for pre- and post-test counselling L La statement that the results of the test might be able to reveal information about genetic relationships L La statement that the results of the genetic test might have implications when purchasing life insurance L La statement that third parties, such as law enforcement agencies, may have access to consumers' biological samples without their consent if laws exist that would permit this L Linformation about specific procedures that might need to be followed if the test is to be used for official purposes, such as certain chains of evidence that might need to be maintained in some jurisdictions, if the test is to be used in the courts of law LLa statement that taking DNA from someone else without their consent is generally ethically inappropriate and is a criminal offence in some jurisdictionsLLinformation about what will happen to consumers' biological samples, and personal and genetic data, if the company ceases trading**4.2** The test provider should provide information to consumers about the association between a genetic variant and a disease, condition or trait for each genetic test that they offer in a format that is easy to understand.**4.3** The test provider should make available to consumers, information about the scope of the test, its accuracy and limitations. Information about the analytical and clinical validity* of each of the genetic markers used in the test should be made available. Other factors, such as behaviour or environmental conditions, that will play a role in determining the development of the condition or trait under investigation should be listed.**4.4** The test provider should provide information about the likely outcomes of the genetic test and the decisions that a consumer may face after taking the test. They should also identify prospectively any likely further investigations that a	Manner which test results will be provided Information about any requirement for genetic counselling Statement that the results may be able to reveal about genetic relationships Statement that the results might have implications for purchasing life insurance State that third parties (e.g. law enforcement) may have access to samples if required by law Statement that taking DNA from someone else is -ethically inappropriate -a criminal offence in some jurisdictions If company ceases trading or is sold -fate of samples -fate of genetic data -fate of personal data Information about the association between a genetic variant and a disease, condition or trait. Scope of test Accuracy of test Limitations of test Analytical validity of markers Clinical validity of markers Behaviour that will influence

		consumer or member of their family may wish to pursue after receiving the test results.**4.5** If a test provider intends to use a consumer's biological samples and/or associated personal or genetic data for research purposes, the consumer should be informed whether the research has been approved by a research ethics committee or other competent authority, whether the biological sample and data will be transferred to or kept in a biobank or database, and about measures to ensure the security of the sample. The consumer should be informed of any risks or potential benefits associated with participating in the research and whether they will receive feedback on research findings that relate to them (see Principle 6.6).**4.6** If a test provider intends to use the results of a genetic test to make a recommendation to a consumer to purchase a therapeutic product, such as a nutritional agent or supplement, the test provider should make available information about the link between the genetic test result and the efficacy of the indicated product. The test provider should also provide information about other lifestyle choices and behavioural modifications that are known to have a preventative or therapeutic value in relation to the trait linked to the genetic markers tested and whether the consumer can purchase the recommended therapeutic product elsewhere.**4.7** Where the test result indicates that the consumer may benefit from an alteration in the dosage of a medicine, or from an alternative medicine to one currently being taken, the test provider should make available information about the link between the genetic test result and the metabolism of the indicated medicines (see Principles 3.1 and 11.3).**4.8** The test provider should make it clear how and whether a consumer can receive updated test results as part of the service they supply.**4.9**	development of condition Environmental factors that will influence development of condition. Likely outcomes of test Decisions consumer may face Any likely further investigations they may wish to pursue after results. If sample or information will be used for research then -whether approved by ethics committee or similar -whether sample will be transferred to a biobank -Whether sample will be kept in a biobank -Whether data will be transferred to a database -Whether data will be kept in a database -Security measures -Potential risks -Potential benefits -Whether there will be feedback If change in medicine recommended by results then provide information on the link. Will consumers be provided with updated test results Recommendations for appropriate actions based on results

		Where appropriate, outside the context of a consultation with a suitably qualified health professional, the test provider should inform consumers about recommendations or known actions that may help the consumer to take informed decisions about their health or welfare in the light of the test results, including informed interaction with the health care system. **4.10** Where appropriate, the test provider should supply consumers with information about health professionals who are able to offer further advice or support. **4.11** For tests in categories 1–6, an appropriately qualified professional, with recognised training and qualifications, employed by or representing the test provider, who is regulated by an appropriate professional body, should be responsible for ensuring that consumers are provided with all of the information specified in this section of the Principles. This requirement should apply to tests in other categories where similar professional structures exist. The test provider should require consumers to sign a statement confirming that they give their informed consent to the specific genetic tests to be undertaken on their biological material. The document should record the sample provider's age and that they have read and understood the information with which they have been provided. The statement should include an explanation of what will happen to the consumer's biological samples and personal data if the controlling share of the company is taken over by a third party. Companies offering direct-to-consumer genetic tests should be aware of the laws that exist in some countries prohibiting DNA theft, which make it illegal to obtain or test DNA without the consent of the person from whom it originated. In line with these laws a test provider should make consumers aware of the law and	Appropriate information about useful health professionals Significance of results Recommend that consumers should not alter medicine based on results but take to doctor State when results can only give relative, rather than absolute, risk If option of sending results by email then state not secure

		should not perform a test if they have reason to believe that a biological sample they have been provided with for genetic testing purposes has been taken from a third party who has not given their consent for the tests to be performed. Requests to recover DNA for genetic testing purposes from secondary objects or materials, when there is reason to believe that the person from whom the DNA originates is still alive, should raise suspicion and should be declined. **Provision of results11.1** The results of genetic tests and the significance that should be attributed to a particular genetic test result should be described to the consumer in a format that is easy to understand.**11.2** When testing for a condition or trait, where such conditions or traits are determined, at least in part, by other, non-genetic factors in addition to genetic markers, the test provider should make consumers aware of these other factors when providing results of genetic tests. In addition, the test provider should supply an indication of the level of significance that an individual should attribute to the genetic test results in comparison with the significance of these other factors, and this should be provided to the consumer in a format that is easy to understand.**11.3** When providing consumers with the test results for tests in category 6 (pharmacogenetic tests), the test provider should strongly recommend that the consumer does not alter the dosage of any existing medication on the basis of the test results and to take the results of the pharmacogenetic test to a medical practitioner for personalised interpretation of the test result. The test provider should give the consumer appropriate information to take with them to their medical practitioner to aid the interpretation of the test results.**11.4** The test provider should take care not to	

		overstate the value or significance of the results of the genetic test when providing the test results.**11.5** The test provider should state clearly when a genetic test result can only give an indication of relative risk in relation to the general population as opposed to an absolute risk, bearing in mind that either might only be calculable in the context of a family history analysis.**11.6** The test provider should have in place a process to evaluate how well consumers are able to understand the background information and test results they have received, and take steps to improve their information and results provision in accordance with the findings.**11.7** The test provider should ensure that the provision of genetic test results is undertaken in such a way as to retain the confidentiality of personal and genetic data. When genetic test results are provided electronically, the test provider should ensure that appropriate security measures are in place to maintain the confidentiality of data transmitted. If the option of sending test results via email is offered by the test provider, consumers should be made aware that this method is generally not secure. For tests in categories 1–6 (and categories 7 and 8 where these have been evaluated as 'high impact' – see 'How to use the Principles') the test provider should be able to provide consumers, at the time of testing or at any subsequent stage, with information about opportunities that are available for any further consultation with health professionals.	

Appendix F Items Removed from Content Analysis

"Sensibility of the test" was removed as unclear as to what the meaning was.

"Claims about clinical validity should be supported with an appropriate reference" was removed as unclear whether as to exact meaning – it may have meant references for evidence for genetic associations, which was covered by several other items. Alternatively, it may have been to do with the clinical use of the test, and all of the websites emphasise that the tests are for informational rather than medical purposes.

"Objectivity of analysis" was removed as it is too obvious an item to expect information on, as tests are obviously not subjective.

"Availability of cancer genetic risk assessment" was removed as unclear to meaning.

"Possibility of following the "IBD" segment containing a specific gene" was removed as to do with ancestry rather than health.

Appendix G Identification of Providers of DTC Genetic Tests

Company	Suitable	Reason if not	Source	Website
23andme	Yes		GPPC	https://www.23andme.com/
Advanced Healthcare, Inc.	No	No health tests	GPPC	http://www.advanceddna.in/
AIBioTech	No	Research tests focus not DTC	GPPC	http://www.aibiotech.com/
Atlas Sports Genetics	No	Sports testing only	GPPC	http://www.atlasgene.com/
Athleticode	No	Predisposition to sports injuries only	GPPC	http://athleticode.com/
deCODE Genetics	Yes		GPPC	https://www.decodeme.com
DNA-CARDIOCHECK	No	Only examines thrombosis	GPPC	http://www.dnaidcheck.com/whatis.html
easyDNA	Yes		GPPC	http://www.easy-dna.com/ (plus other country endings eg. .co.uk)
EnteroLab	No	Only examines gluten sensitivity	GPPC	http://www.enterolab.com/
GenePlanet	Yes		GPPC	http://www.geneplanet.com/
Genetic Testing Laboratories (GTL)	No	No health tests	GPPC	http://www.gtldna.co.uk/
Graceful Earth	No	Only examines Alzheimer's disease	GPPC	http://gracefulearth.com/
HealthCheck USA	No	Only examines one health condition per test	GPPC	http://www.healthcheckusa.com/
Concept Holistic Health	No	Testing at a clinic, not ordering a test over the internet	GPPC	http://www.conceptholistichealth.com.au/
Inherent Health/Interleuken Genetics, Inc.	Yes		GPPC	http://www.inherenthealth.com/
Lumigenix	Yes		GPPC	https://www.lumigenix.com/

Map My Gene	Yes		GPPC	http://www.mapmygene.com/
Test Country	Yes		GPPC	http://www.testcountry.com/ (plus local ones)
Viaguard/Accu-metrics	Yes		GPPC	http://www.accu-metrics.com/
vuGene	No	Individual tests for children's sleep apnea, stroke and Alzheimer's only	GPPC	http://www.mygenesdirect.com/
Genelex	No	Pharmacogenomic	GPPC	http://www.healthanddna.com/
Illumina	No	Not DTC. Only whole genome sequencing, doesn't provide risks for diseases	GPPC	http://www.illumina.com/
Knome	No	Not DTC, not aimed at consumers	GPPC	http://www.knome.com/
Navigenics	Yes (not DTC)	No longer accepting orders from 3rd August 2012	GPPC	http://www.navigenics.com/
Pathway Genomics	Yes (not DTC)		GPPC	https://www.pathway.com/
Pediatrix Medical Group	No	Only tests newborns for hearing loss susceptibility	GPPC	http://www.pediatrix.com/
Perkin Elmer Genetics	No	Metabolic disorders only	GPPC	http://www.perkinelmergenetics.com/
International Biosciences	Yes		Search terms from GPPC	http://www.ibdna.com/regions/UK/EN/?page=home
Genetic Health	Yes		Search terms from GPPC	http://www.genetic-health.co.uk/index.asp

Appendix H Data Tables for Statistical Tests

Tables for Statistical Tests Reported in Section 5.3

Differences in Health Behaviours and Anxiety Levels

Health-Behaviour Scores

All Respondents

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Health_Behaviour_Score is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.022	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Respondent_Group * High_or_Low Crosstabulation

Count

		High_or_Low		Total
		1.00	2.00	
Respondent_Group	Consumers	42	110	152
	Potential Consumers	32	32	64
Total		74	142	216

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	10.005[a]	1	.002		
Continuity Correction[b]	9.037	1	.003		
Likelihood Ratio	9.751	1	.002		
Fisher's Exact Test				.003	.001
Linear-by-Linear Association	9.959	1	.002		
N of Valid Cases	216				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 21.93.

b. Computed only for a 2x2 table

Respondents who Purchased for Genealogical Reasons Excluded

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Health_Behaviour_Score is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.030	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Respondent_Group * Low_or_High Crosstabulation

Count

		Low_or_High		Total
		1.00	2.00	
Respondent_Group	Consumers	31	85	116
	Potential Consumers	29	29	58
Total		60	114	174

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	9.272[a]	1	.002		
Continuity Correction[b]	8.271	1	.004		
Likelihood Ratio	9.097	1	.003		
Fisher's Exact Test				.004	.002
Linear-by-Linear Association	9.219	1	.002		
N of Valid Cases	174				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 20.00.

b. Computed only for a 2x2 table

Individual Health Behaviours

All Respondents

Salt Intake

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	62	109	171
	Potential Consumers	31	38	69
Total		93	147	240

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	1.557[a]	1	.212		
Continuity Correction[b]	1.213	1	.271		
Likelihood Ratio	1.543	1	.214		
Fisher's Exact Test				.242	.136
Linear-by-Linear Association	1.551	1	.213		
N of Valid Cases	240				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 26.74.

b. Computed only for a 2x2 table

Fat Intake

Respondent_Group * Not_Followng_Or_Following Crosstabulation

Count

		Not_Followng_Or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	76	101	177
	Potential Consumers	36	37	73
Total		112	138	250

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	.850[a]	1	.357		
Continuity Correction[b]	.612	1	.434		
Likelihood Ratio	.848	1	.357		
Fisher's Exact Test				.402	.217
Linear-by-Linear Association	.847	1	.358		
N of Valid Cases	250				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 32.70.

b. Computed only for a 2x2 table

Fibre Intake

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	44	125	169
	Potential Consumers	27	49	76
Total		71	174	245

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	2.294[a]	1	.130		
Continuity Correction[b]	1.856	1	.173		
Likelihood Ratio	2.248	1	.134		
Fisher's Exact Test				.170	.087
Linear-by-Linear Association	2.285	1	.131		
N of Valid Cases	245				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 22.02.

b. Computed only for a 2x2 table

Fruit and Vegetables

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	84	97	181
	Potential Consumers	49	31	80
Total		133	128	261

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	4.890[a]	1	.027		
Continuity Correction[b]	4.314	1	.038		
Likelihood Ratio	4.924	1	.026		
Fisher's Exact Test				.032	.019
Linear-by-Linear Association	4.871	1	.027		
N of Valid Cases	261				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 39.23.

b. Computed only for a 2x2 table

Smoking

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	13	166	179
	Potential Consumers	5	76	81
Total		18	242	260

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	.103[a]	1	.749		
Continuity Correction[b]	.003	1	.955		
Likelihood Ratio	.105	1	.746		
Fisher's Exact Test				1.000	.489
Linear-by-Linear Association	.102	1	.749		
N of Valid Cases	260				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 5.61.

b. Computed only for a 2x2 table

Exercise

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	58	114	172
	Potential Consumers	30	47	77
Total		88	161	249

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	.639[a]	1	.424		
Continuity Correction[b]	.430	1	.512		
Likelihood Ratio	.634	1	.426		
Fisher's Exact Test				.474	.255
Linear-by-Linear Association	.637	1	.425		
N of Valid Cases	249				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 27.21.

b. Computed only for a 2x2 table

Respondents who Purchased for Genealogical Reasons Excluded

Salt Intake

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	47	84	131
	Potential Consumers	28	35	63
Total		75	119	194

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	1.316[a]	1	.251		
Continuity Correction[b]	.980	1	.322		
Likelihood Ratio	1.307	1	.253		
Fisher's Exact Test				.273	.161
Linear-by-Linear Association	1.310	1	.252		
N of Valid Cases	194				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 24.36.

b. Computed only for a 2x2 table

Fat Intake

Resondent_Group * Not_Following_or_Followin Crosstabulation

Count

		Not_Following_or_Followin		Total
		Not Following	Following	
Resondent_Group	Consumers	57	78	135
	Potential Consumers	34	32	66
Total		91	110	201

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	1.545[a]	1	.214		
Continuity Correction[b]	1.193	1	.275		
Likelihood Ratio	1.542	1	.214		
Fisher's Exact Test				.230	.137
Linear-by-Linear Association	1.537	1	.215		
N of Valid Cases	201				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 29.88.

b. Computed only for a 2x2 table

Fibre Intake

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	32	99	131
	Potential Consumers	23	46	69
Total		55	145	200

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	1.798[a]	1	.180		
Continuity Correction[b]	1.379	1	.240		
Likelihood Ratio	1.768	1	.184		
Fisher's Exact Test				.187	.121
Linear-by-Linear Association	1.789	1	.181		
N of Valid Cases	200				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 18.98.

b. Computed only for a 2x2 table

Fruit and Vegetables

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	65	74	139
	Potential Consumers	46	27	73
Total		111	101	212

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	5.068[a]	1	.024		
Continuity Correction[b]	4.437	1	.035		
Likelihood Ratio	5.114	1	.024		
Fisher's Exact Test				.030	.017
Linear-by-Linear Association	5.044	1	.025		
N of Valid Cases	212				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 34.78.

b. Computed only for a 2x2 table

Smoking

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	8	128	136
	Potential Consumers	5	69	74
Total		13	197	210

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	.063[a]	1	.802		
Continuity Correction[b]	.000	1	1.000		
Likelihood Ratio	.062	1	.803		
Fisher's Exact Test				.773	.509
Linear-by-Linear Association	.063	1	.802		
N of Valid Cases	210				

a. 1 cells (25.0%) have expected count less than 5. The minimum expected count is 4.58.

b. Computed only for a 2x2 table

Exercise

Respondent_Group * Not_Following_or_Following Crosstabulation

Count

		Not_Following_or_Following		Total
		Not Following	Following	
Respondent_Group	Consumers	37	93	130
	Potential Consumers	27	43	70
Total		64	136	200

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Pearson Chi-Square	2.137[a]	1	.144		
Continuity Correction[b]	1.698	1	.193		
Likelihood Ratio	2.109	1	.146		
Fisher's Exact Test				.155	.097
Linear-by-Linear Association	2.127	1	.145		
N of Valid Cases	200				

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 22.40.

b. Computed only for a 2x2 table

Health Anxiety

Anxiety in General

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of General_Anxiety is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.629	Retain the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Anxiety about Serious Disease

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Anxiety_Serious_Disease is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.301	Retain the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Potential Consumers Group

Group Statistics

	Respondent_Group	N	Mean	Std. Deviation	Std. Error Mean
Behaviour_Score	'True' Potential Consumers	45	3.67	1.365	.204
	Not Received	19	3.58	1.346	.309

Independent Samples Test

		Levene's Test for Equality of Variances		t-test for Equality of Means						
		F	Sig.	t	df	Sig. (2-tailed)	Mean Difference	Std. Error Difference	95% Confidence Interval of the Difference	
									Lower	Upper
Behaviour_Score	Equal variances assumed	.061	.806	.236	62	.814	.088	.372	-.656	.831
	Equal variances not assumed			.237	34.368	.814	.088	.370	-.664	.839

Perceived Health Risk

Perceived Health Risk and Health-Behaviour Score

Correlations

			Perceived_Disease_Risk	Behaviour_Score
Spearman's rho	Perceived_Disease_Risk	Correlation Coefficient	1.000	-.077
		Sig. (2-tailed)	.	.364
		N	141	141
	Behaviour_Score	Correlation Coefficient	-.077	1.000
		Sig. (2-tailed)	.364	.
		N	141	141

Perceived Health Risk and Anxiety in General

Correlations

			Perceived_Disease_Risk	Anxiety_General
Spearman's rho	Perceived_Disease_Risk	Correlation Coefficient	1.000	.100
		Sig. (2-tailed)	.	.203
		N	165	165
	Anxiety_General	Correlation Coefficient	.100	1.000
		Sig. (2-tailed)	.203	.
		N	165	165

Perceived Health Risk and Anxiety about Serious Disease

Correlations

			Perceived_Disease_Risk	Anxiety_Serious_Disease
Spearman's rho	Perceived_Disease_Risk	Correlation Coefficient	1.000	.147
		Sig. (2-tailed)	.	.059
		N	165	165
	Anxiety_Serious_Disease	Correlation Coefficient	.147	1.000
		Sig. (2-tailed)	.059	.
		N	165	165

Perceived Health Risk and Change in Behaviour

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Perceived_Disease_Risk is the same across categories of Behaviour_Change.	Independent-Samples Kolmogorov-Smirnov Test	.778	Retain the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Underlying Factors

Difference between Consumers and Potential Consumers

Age Group

Respondent_Group * Age Crosstabulation

Count

		Age				Total
		18-29	30-44	45-60	60+	
Respondent_Group	Consumers	31	51	41	28	151
	Potential Consumers	26	24	10	3	63
Total		57	75	51	31	214

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)	Point Probability
Pearson Chi-Square	15.617[a]	3	.001	.001		
Likelihood Ratio	16.581	3	.001	.001		
Fisher's Exact Test	15.750			.001		
Linear-by-Linear Association	15.465[b]	1	.000	.000	.000	.00
N of Valid Cases	214					

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 9.13.

b. The standardized statistic is -3.933.

Correlations

			Age	Behaviour_Score
Spearman's rho	Age	Correlation Coefficient	1.000	.130
		Sig. (2-tailed)	.	.057
		N	214	214
	Behaviour_Score	Correlation Coefficient	.130	1.000
		Sig. (2-tailed)	.057	.
		N	214	214

Proportion in Managerial and Professional Occupations Socioeconomic Category

Respondent_Group * Socioeconomic_Category Crosstabulation

Count

		Socioeconomic_Category		Total
		Managerial and Professional Occupations	Other Categories	
Respondent_Group	Consumers	128	20	148
	Potential Consumers	46	15	61
Total		174	35	209

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)	Point Probability
Pearson Chi-Square	3.801[a]	1	.051	.066	.043	
Continuity Correction[b]	3.048	1	.081			
Likelihood Ratio	3.594	1	.058	.066	.043	
Fisher's Exact Test				.066	.043	
Linear-by-Linear Association	3.783[c]	1	.052	.066	.043	.025
N of Valid Cases	209					

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 10.22.

b. Computed only for a 2x2 table

c. The standardized statistic is 1.945.

Gender

Respondent_Group * Gender Crosstabulation

Count

		Gender		Total
		Female	Male	
Respondent_Group	Consumers	57	93	150
	Potential Consumers	26	38	64
Total		83	131	214

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)	Point Probability
Pearson Chi-Square	.130[a]	1	.718	.760	.416	
Continuity Correction[b]	.043	1	.836			
Likelihood Ratio	.130	1	.719	.760	.416	
Fisher's Exact Test				.760	.416	
Linear-by-Linear Association	.130[c]	1	.719	.760	.416	.11
N of Valid Cases	214					

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 24.82.

b. Computed only for a 2x2 table

c. The standardized statistic is -.360.

Proportion of Caucasian Ethnicity

Respondent_Group * Ethnicity Crosstabulation

Count

		Ethnicity		Total
		Caucasian	Other Ethnicities	
Respondent_Group	Consumers	124	22	146
	Potential Consumers	50	9	59
Total		174	31	205

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)	Point Probability
Pearson Chi-Square	.001[a]	1	.973	1.000	.564	
Continuity Correction[b]	.000	1	1.000			
Likelihood Ratio	.001	1	.973	1.000	.564	
Fisher's Exact Test				1.000	.564	
Linear-by-Linear Association	.001[c]	1	.973	1.000	.564	.170
N of Valid Cases	205					

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 8.92.

b. Computed only for a 2x2 table

c. The standardized statistic is .034.

Level of Education

Respondent_Group * Level_of_Education Crosstabulation

Count

		Level_of_Education				Total
		School	Post-school	Bachelor's Degree	Postgraduate Degree	
Respondent_Group	Consumers	3	3	60	83	149
	Potential Consumers	3	4	26	31	64
Total		6	7	86	114	213

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)	Point Probability
Pearson Chi-Square	4.025[a]	3	.259	.271		
Likelihood Ratio	3.695	3	.296	.364		
Fisher's Exact Test	4.131			.236		
Linear-by-Linear Association	2.626[b]	1	.105	.108	.067	.023
N of Valid Cases	213					

a. 4 cells (50.0%) have expected count less than 5. The minimum expected count is 1.80.

b. The standardized statistic is -1.621.

Proportion Resident in USA

Respondent_Group * Country_of_Residence Crosstabulation

Count

		Country_of_Residence		Total
		Other Countries	USA	
Respondent_Group	Consumers	37	114	151
	Potential Consumers	22	42	64
Total		59	156	215

Chi-Square Tests

	Value	df	Asymp. Sig. (2-sided)	Exact Sig. (2-sided)	Exact Sig. (1-sided)	Point Probability
Pearson Chi-Square	2.200[a]	1	.138	.181	.095	
Continuity Correction[b]	1.732	1	.188			
Likelihood Ratio	2.146	1	.143	.181	.095	
Fisher's Exact Test				.181	.095	
Linear-by-Linear Association	2.190[c]	1	.139	.181	.095	.044
N of Valid Cases	215					

a. 0 cells (0.0%) have expected count less than 5. The minimum expected count is 17.56.

b. Computed only for a 2x2 table

c. The standardized statistic is -1.480.

Weighted Health-Behaviour Scores

Age

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Weighted_Behaviour_Score is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.034	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Socioeconomic Category

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Weighted_Behaviour_Score is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.037	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Ethnicity

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Weighted_Behaviour_Score is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.048	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Level of Education

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Weighted_Behaviour_Score is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.028	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Gender

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Weighted_Behaviour_Scores is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.016	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Country of Residence

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Weighted_Behaviour_Score is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.006	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Stepwise Multiple Regression

Model Summary

Model	R	R Square	Adjusted R Square	Std. Error of the Estimate
1	.157[a]	.025	.020	1.383

a. Predictors: (Constant), Respondent_Group

ANOVA[a]

Model		Sum of Squares	df	Mean Square	F	Sig.
1	Regression	9.204	1	9.204	4.810	.030[b]
	Residual	363.541	190	1.913		
	Total	372.745	191			

a. Dependent Variable: Behaviour_Score

b. Predictors: (Constant), Respondent_Group

Coefficients[a]

Model		Unstandardized Coefficients		Standardized Coefficients	t	Sig.
		B	Std. Error	Beta		
1	(Constant)	4.659	.301		15.475	.000
	Respondent_Group	-.484	.221	-.157	-2.193	.030

a. Dependent Variable: Behaviour_Score

Excluded Variables[a]

Model		Beta In	t	Sig.	Partial Correlation	Collinearity Statistics
						Tolerance
1	Gender	-.136[b]	-1.905	.058	-.137	1.000
	Age_Group	.032[b]	.426	.670	.031	.922
	Residence_in_USA	-.062[b]	-.858	.392	-.062	.989
	Caucasian_Ethnicity	.053[b]	.732	.465	.053	1.000
	Education_Level	.121[b]	1.691	.093	.122	.993
	Socioeconomic_Category_One	-.078[b]	-1.086	.279	-.079	.990

a. Dependent Variable: Behaviour_Score

b. Predictors in the Model: (Constant), Respondent_Group

Mean Website Assessment Scores

Mean Website Assessment Score and Health-Behaviour Score

Correlations

			Behaviour_Score	Mean_Website_Assessment_Score
Spearman's rho	Behaviour_Score	Correlation Coefficient	1.000	.142
		Sig. (2-tailed)	.	.084
		N	149	149
	Mean_Website_Assessment_Score	Correlation Coefficient	.142	1.000
		Sig. (2-tailed)	.084	.
		N	149	180

Mean Website Assessment Score and Anxiety about Serious Disease

Correlations

			Mean_Website_Assessment_Score	Anxiety_Serious_Disease
Spearman's rho	Mean_Website_Assessment_Score	Correlation Coefficient	1.000	-.147
		Sig. (2-tailed)	.	.051
		N	180	176
	Anxiety_Serious_Disease	Correlation Coefficient	-.147	1.000
		Sig. (2-tailed)	.051	.
		N	176	176

Mean Website Assessment Score and Anxiety in General

Correlations

			Mean_Website_Assessment_Score	Anxiety_General
Spearman's rho	Mean_Website_Assessment_Score	Correlation Coefficient	1.000	-.155*
		Sig. (2-tailed)	.	.040
		N	180	176
	Anxiety_General	Correlation Coefficient	-.155*	1.000
		Sig. (2-tailed)	.040	.
		N	176	176

*. Correlation is significant at the 0.05 level (2-tailed).

Mean Website Assessment Score and Behaviour Change

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Mean_Website_Assessment_Score is the same across categories of Change_in_Behaviour.	Independent-Samples Kolmogorov-Smirnov Test	.451	Retain the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Tables for Statistical Tests Reported in Section 5.4

'I am generally satisfied with the information provided on the website'

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Generally_Satisfied is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.004	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

'I had trouble understanding some of the information on the website'

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Trouble_Understanding is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	1.000	Retain the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

'There is adequate information on the website to make a decision about buying a test'

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Adequate_Information is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.013	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

‘The information on the website appears to be trustworthy’

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Trustworthy is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.050	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

‘The information on the website appears to be reliable’

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Reliable is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.031	Reject the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

‘I had to look at other sources to find enough information to make a decision about buying a test’

Hypothesis Test Summary

	Null Hypothesis	Test	Sig.	Decision
1	The distribution of Other_Sources is the same across categories of Respondent_Group.	Independent-Samples Kolmogorov-Smirnov Test	.488	Retain the null hypothesis.

Asymptotic significances are displayed. The significance level is .05.

Appendix I Explanation of Category Names

Category Names in Section 5.2.1

Category	Description
Blogs	Refers to the number of people who looked at blogs. The 'specifically scientific' subcategory refers to people who have looked at blogs that they specifically mentioned (or are well known) as science blogs, genetics blogs, blogs specifically about DTC tests, blogs by experts etc. The 'non-specific' category refers to the number of people who looked at blogs that are not specifically mentioned or well known as science blogs etc.
Google	Refers to the number of people who performed a Google search, a search by a different search engine or just a general internet search.
General internet use	Refers to the number of people who only mentioned general non-specific internet use.
Articles	Refers to the number of people who looked at articles about DTC genetic tests, including both online and offline articles.
Literature	Refers to the number of people who looked at the scientific literature, or searched through a scientific database or Google Scholar.
DTC website	Refers to the number of people who mentioned looking at the DTC genetic testing company website. The 'comprehensive look' subcategory refers to those who specifically stated that they used the resources or references provided on the website. The 'unspecified' category refers to those who did not mention doing so.
Forums	Refers to the number of people who looked at internet forums. The 'specifically scientific' subcategory refers to the number of people who looked at forums that they specifically mentioned (or are well known) as science forums, genetics forums, forums specifically about DTC genetic tests etc. The 'non-specific' subcategory refers to the number of people who looked at forums that were not specifically mentioned or are well known as science forums etc.
General friends	Refers to the number of people who mentioned generally talking to friends, colleagues, relatives etc. or something to do with word of mouth.
Friends who are experts	Refers to the number of people who talked to friends, colleagues, relatives, etc. who are explicitly stated as knowing a lot about the field, or who are doctors (including their personal doctor).
Reviews	Refers to the number of people who read reviews of the tests.
Mailing Lists	Refers to the number of people who looked at mailing lists. The 'specifically scientific' subcategory refers to the number of people who looked at mailing lists that they specifically mentioned (or are well known) as science lists, genetics lists, lists specifically about DTC genetic tests etc. The 'non-specific' subcategory refers to the number of people who looked at mailing lists that were not specifically mentioned or well known as science lists etc.
Friends who have taken the test	Refers to the number of people who have talked to friends, colleagues, relatives etc. who have taken a DTC genetic test.
DTC company	Refers to the number of people who mentioned looking at the DTC genetic testing company, or which companies they were interested in, but who did not specifically mention looking at the website.
Wikipedia	Refers to the number of people who looked at Wikipedia
Other named website	Refers to the number of people who looked at a specific website that is not covered elsewhere.
General reports	Refers to people who looked at general reports or feedback of people who have

	taken a test, or people's accounts in blogs, users ratings etc
Twitter	Refers to the number of people who looked at Twitter
Previous testing	Refers to the number of people who mentioned that they have been previously tested.
Books	Refers to the number of people who looked at books for information.
Experts not in the person	Refers to the number of people who mentioned experts who they did not talk to in person e.g. TV doctors or scientists etc., an expert in an interview etc.
Workshop	Refers to the number of people who attended a workshop or conference presentation with information about the tests.
Work	Refers to the number of people who mentioned work or something work-related.
Magazines	Refers to the number of people who looked at magazines or other media.
ISOGG	Refers to the number of people who mentioned the International Society of Genetic Genealogy.
General reading	Refers to the number of people who mentioned general reading.
Documentary	Refers to the number of people who watched a documentary about the tests.
Online support groups	Refers to the number of people who looked at online support groups.
Podcasts	Refers to the number of people who looked at podcasts.
YouTube	Refers to the number of people who looked at YouTube.
Non-specific radio interview	Refers to the number of people who mentioned a radio interview without specifying who was interviewed.
Other sources	Refers to the number of people who looked at other sources of information.

Category Names in Section 5.5.1

Category	Explanation
Coverage	Something to do with the coverage of the test, such as the number of SNPs (mutations) tested, what information is analysed or what this means.
Cost	Cost of the tests.
Tests	Something to do with what tests are available, who gives the best service, what is the most common test etc.
Reliability	Something to do with the reliability of the tests.
Reviews	Reviews, feedback or testimonials about the tests.
Information produced by tests	Looking for what information is produced by the tests, what is the nature of the information provided etc.
Usefulness	Something about the how useful the tests are.
Technical details of analysis	Something to do with the technical aspects of the analysis, such as what chip is used, what the methods of analysis are etc.
Raw data or third party analysis	Information about access to raw data and/or the interpretation of raw data by third party software.
Sample reports	A sample of what the results and report look like.
Bibliography	The literature used by the companies and what the tests are based on.
Who is associated	Looking to see if any well-known people are associated with the tests.
Information available elsewhere	Looking to see what information about the area is available from sources other than the DTC genetic testing companies.
Existing data	Looking for any existing data about the tests.
Interpretation	Looking to see if there is any information about interpreting the

	results.
Learned so far	Information about what has been learned so far from the tests.
Genetics of a specific disease	Something to do with the genetics of a specific disease, such as diabetes.
DTC genetic tests in general	Generally looking for information about the tests.
Ancestry information	Information about the ancestry aspect of the tests.
Publishing records of scientists	Looking for the publication records of the scientists involved in the tests.
Method	Information about the methods the tests use.
Support	Something to do with customer support, commitment to customers, ability to answer questions etc.
Company history	Information about the history of the company.

Category Names in Section 5.6.1

Category	Explanation
Coverage	Something to do with the coverage of the test, such as the number of SNPs (mutations) tested, what information is analysed or what this means.
Accuracy	Something to do with the accuracy, reliability, rate of errors, possibility of misinterpretation, scientific credibility, quality or repeatability of the test.
Cost	Cost of the test.
Privacy	Something to do with privacy or who details (e.g. results) are shared with (this excludes named details though, which come under confidentiality).
Results	Something to do with how detailed the results are, what form they come in, what you can learn from them, what applications they might have etc.
Sample	How the sample is collected or how the test is administered/works.
Analysis	Detailed information about the analysis (rather than just how the test works), methodology, technology used, chip or platform used, depth of analysis etc.
Data	Access to raw data or ability to move data.
Trustworthy	Something to do with the trustworthiness of the company, if it has a good reputation, is capable of providing the service, is serious etc.
Interface	Something to do with the user interface or sample results, what can be expected from results, ease of access, tools etc.
Security	Security of the data and how it is stored.
Confidentiality	Confidentiality of personal details and results, and any impact of details of test on health insurance.
Time	Time to wait for results.
Interpretation	Looking if there is help in interpreting results.
Sharing	Ability to share results, search database, find relatives etc.
Updates	Looking if updates will be available.
Useful	Something about the usefulness of the tests.
Lab information	Information about the lab, such as is it properly certified, does it have a good reputation, does it use established procedures etc.
General	General wanting to know lots of information or everything possible.
Company	Looking to see which companies are selling the tests, who are they, who does the testing etc.
Business model	What is the business model of the company.
Sample size	Something to do with the sample size or size of database.
Comparisons	Comparisons between different tests, chips, what SNPs are on the chips etc.
Site founders	Looking for who the founders are, what their expertise is, who owns the

	company etc.
Website	Quality and usability of the website.
Prevention	Looking for information on preventative strategies.
Transparency	Transparency of the tests and process.
Downfalls	Any downfalls to taking the test.
Repeatability	Something to do with the repeatability of the results between different companies.
Years in business	Looking to see how many years the company has been in business.
3rd Party tools	Looking for Information about 3rd party tools for analysing data.
Stability of company	How stable the company is.
Ancestry examples	Examples of how the ancestry parts work.
Users	Looking for information about other users.
Safety	Something about the safety of the test.
Ownership	Looking to see who owns the raw data.
Type of testing	What type of testing is available.
Support	Looking at what user support is available.
Treatment they have	Looking for information about treatments they sell.

Categories	Explanation
Interpretation/research into raw data	Information on how to interpret data, including how to do research on raw data.
Doctor	How to present results to doctor.
Meaning of words	What certain words in the report mean.
Application	What the applications of the results are.
Clinically actionable	If any of the results are clinically actionable.
Gene/environment interaction	Information on gene/environment interactions.
Reuse Sample	Can the sample be reused for future tests.

Appendix J Glossary

Acronym	Definition
DTC	Direct-to-consumer
DNA	Deoxyribonucleic acid
RNA	Ribonucleic acid
CAD	Coronary artery disease
PKU	Phenylketonuria
ACM	Association for computing machinery
FDA	Food and drug administration
NS-SEC	National statistics socio-economic classification
GPPC	Genetics and public policy centre
ISOGG	International society of genetic genealogy
GWAS	Genome-wide association study
ARMD	Age-related macular degeneration
SNP	Single nucleotide polymorphism
CLIA	Clinical laboratory improvement amendments
HIPAA	Health insurance portability & accountability act
STR	Short tandem repeats
IBD	Identical by descent
WGS	Whole genome sequencing
CMIS	Comprehensive model of information seeking