Public perception of genetics in healthcare

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Abstract

The UK is a world leader in genomic medicine, with plans to collect 5 million genomes by 2024. The use of genetic data in healthcare is posited to bring many benefits to UK healthcare.

However, genetics is not widely understood by the public. The deficit model of knowledge suggests that misunderstandings about genetics are likely to be linked to a lack of acceptance of genomic medicine. It is important to characterise the nature of this relationship as this will help to guide mainstreaming of genetic testing into NHS practice. Previous studies examining this relationship show mixed results; the nature of the relationship may depend on the specific type of knowledge studied. Therefore, my first study explored UK participants' biological knowledge of genetics, their clinical knowledge of genomic medicine, and their perception of their genetic knowledge. The results indicated that neither biological nor clinical knowledge predicted participants' acceptance of genomics. However, participants' perception of their own genetic knowledge was found to predict their acceptance. This suggests that to increase acceptance, we should focus our resources on ensuring the public are confident in their knowledge.

My second study was a qualitative exploration of participants' opinions about genomic medicine. Participants were given information about genomic medicine prior to the study. The results indicated that although participants recognised some benefits of genomic medicine, they also had concerns. For example, whilst genetic data were considered to have the ability to empower you to live your life to the full, participants were also concerned about the potential for genetic results to have a detrimental psychological impact. Overall, the results pointed to the importance of genetic counsellors and maintaining anonymity throughout the genetic testing process, as well as showing that the public are keen to get involved with the development of genomic policy.

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Author's declaration

I declare that the work in this dissertation was carried out in accordance with the requirements of the University's *Regulations and Code of Practice for Research Degree Programmes* and that it has not been submitted for any other academic award. Except where indicated by specific reference in the text, the work is the candidate's own work. Work done in collaboration with, or with the assistance of, others, is indicated as such. Any views expressed in the dissertation are those of the author.

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List of Abbreviations

DNA: Deoxyribose Nucleic Acid

EMPAG: European Meeting on Psychosocial Aspects of Genetics

GDPR: General Data Protection Regulation

GMC: Genome Medicine Centre

NHS: National Health Service

NHGRI: National Human Genome Research Institute

PIS: Participant Information Sheets

SES: Socio-economic status

WGS: Whole Genome Sequencing

Chapter 1: Introduction & literature review

1.1 Thesis motivation

My undergraduate dissertation findings indicated a growing interest in the use of genomic medicine within the UK National Health Service (NHS). This triggered my interest in the public's perception of the use of genetic data in healthcare. I wanted to offer a useful contribution as to how we can improve public acceptance of genomic medicine, and deliver an up-to-date insight into public opinion, particularly as the 100,000 Genomes Project was set to finish two months into my MSc. I decided on a mixed methods approach because it would allow for the collection of rich data. It also meant I could focus on developing my skills in both quantitative and qualitative methodology.

1.1.1 Collaboration with West of England Genomic Medicine Centre

Throughout my MSc, I have collaborated with the West of England Genomic Medicine Centre (GMC). This gave me a useful insight into the reality of integrating genomics into healthcare and the difficulties associated. One of these difficulties concerned the consent process for undertaking genetic testing in a clinical setting. The GMC wanted to improve its understanding of current public views so that the development of consent materials could incorporate the opinions of those it would affect. Through the current thesis and additional collaborative work, I hope to help inform their consent process.

1.2 Thesis overview

In this thesis, I aimed to explore the relationship between knowledge of genetics and acceptance of genomics in the NHS. Previous research indicates the specific type of scientific knowledge studied is important when exploring the relationship between knowledge and acceptance (Allum et al. 2008). I aimed to explore three different aspects of knowledge:

biological, i.e. knowledge of genes and how they work in the body, clinical, i.e. an awareness of the current situation of genomic medicine in the UK, and perception of knowledge, i.e. how confident participants are in their understanding of genetics. I am not aware of any studies that have explored clinical knowledge. Instead, previous studies have focused on biological knowledge (Allum et al. 2014; Human Genetics Commission, 2001; Jallinjoa & Aro, 2000), which misses out on real world applications of knowledge. I also conducted a qualitative investigation into public opinion on the use of genetic data in healthcare. I believe this is one of the first studies to study the public's opinion of clinical genetics after informing participants about genomics beforehand; this allowed us to gather opinions that are not based on false information and ideas.

The importance of investigations using members of the public has been demonstrated multiple times (Davies, 2017; Haga & Willard, 2006; Henneman et al. 2013; Samuel & Farsides, 2018); it may uncover gaps in knowledge or sources of uncertainty that can help to develop more effective communication and maximise the probability that policies will generate satisfaction in all stakeholders. Therefore, based on data from the public, this thesis aims to provide a valuable input to the integration of genomics into the NHS.

1.3 The human genome and genomic medicine

A genome is the entire set of genetic material in a cell and encompasses all of the information required to build and maintain an organism (Genetics Home Reference, 2019). Deoxyribonucleic acid (DNA) contains 4 nucleotides bases that pair up: adenine (A) pairs with thymine (T), and cytosine (C) pairs with guanine (G). A human genome has approximately 3 billion of these nucleotide base pairs (National Human Genome Research Institute, 2018).

Whole genome sequencing (WGS) is a technology that allows researchers to decipher the exact order of the letters - A, T, C and G - in a genome (Icahn School of Medicine, 2012). This can: reveal information about an individual's risk of disease, help to give an accurate diagnosis for a pre-existing condition, or indicate the most effective choice of medical treatment (NHGRI, 2018). This has been labelled 'genomic medicine'.

Globally, the NHS is the single largest integrated healthcare system (NHS England, 2018a), i.e. NHS organisations collaborate with external organisations and benefit from their expertise to deliver high quality healthcare (NHS England, n.d). Therefore, the NHS (2018a) recognises the potential for genomics not only to benefit patients but, through collaboration with the life sciences industry, propel the UK's understanding of disease and its ability to develop medical tools for early detection and effective treatment.

1.4 History of genomic medicine

In 1990, sequencing the human genome took 13 years of research and cost, at minimum, $500 million (National Human Genome Research Institute, 2019). In 2012, the 100,000 Genomes Project was launched, which was the first genomic project to focus on the integration of genomic data into healthcare (PHG Foundation, 2012). Today, we can sequence a genome in a single day for £700 or less (Turnbull et al. 2018) and the UK has just launched the world's first genomic medicine service (Genetic Alliance UK, 2018). To illustrate the speed at which our knowledge of genomics has developed, I have outlined a timeline of the last 30 years of genomic sequencing and its use in healthcare (Figure 1). Further detail of the significant time points can be found below.

- **1990:** The Human Genome Project officially began. The National Human Genome Research Institute recognised this was an international effort to identify the entire sequence of nucleotide base pairs in human DNA (NHGRI, 2013).

- **2003:** Two years earlier than anticipated, the Human Genome Project was completed. Due to limited technology, the finished sequence had a few small gaps however consisted of 99% of the genome and was 99.99% accurate (NHGRI, 2014).

- **2008:** Another international collaboration was launched: 1,000 Genomes Project. The aim was to identify genetic variants, i.e. changes in the most common sequence of nucleotide bases in DNA, with frequencies of ≥1% from multiple populations (1000 Genomes Project Consortium, 2015).

- **2012:** The UK Government announced a 3-5 year plan to sequence 100,000 genomes from patients with a rare disease or cancer (PHG Foundation, 2012). This project shifted genomic sequencing into the medical world.

- **2015: The completion of the 1,000 Genomes Project** (1000 Genomes Project Consortium, 2015) provided a useful and publicly available resource of human genetic variation and reduced sequencing costs (International Genome Sample Resource, 2018).

- **2018:** The completion of transformational 100,000 Genomes Project helped to diagnose approximately 25% of the participants with a rare disease, delivered actionable information for some cancer patients, and provided a vast store of genomic and clinical data for research (Genomics England, 2018).

  In October 2018, the NHS genomic medicine service was initiated. This service aims to integrate WGS into the mainstream care of patients with a rare inherited disease or cancer (Turnbull et al. 2018). Further, to enable a more systematic approach to genomic medicine, the NHS has created a National Genomic Test Directory that details the type and eligibility criteria of available tests (NHS England, 2019).

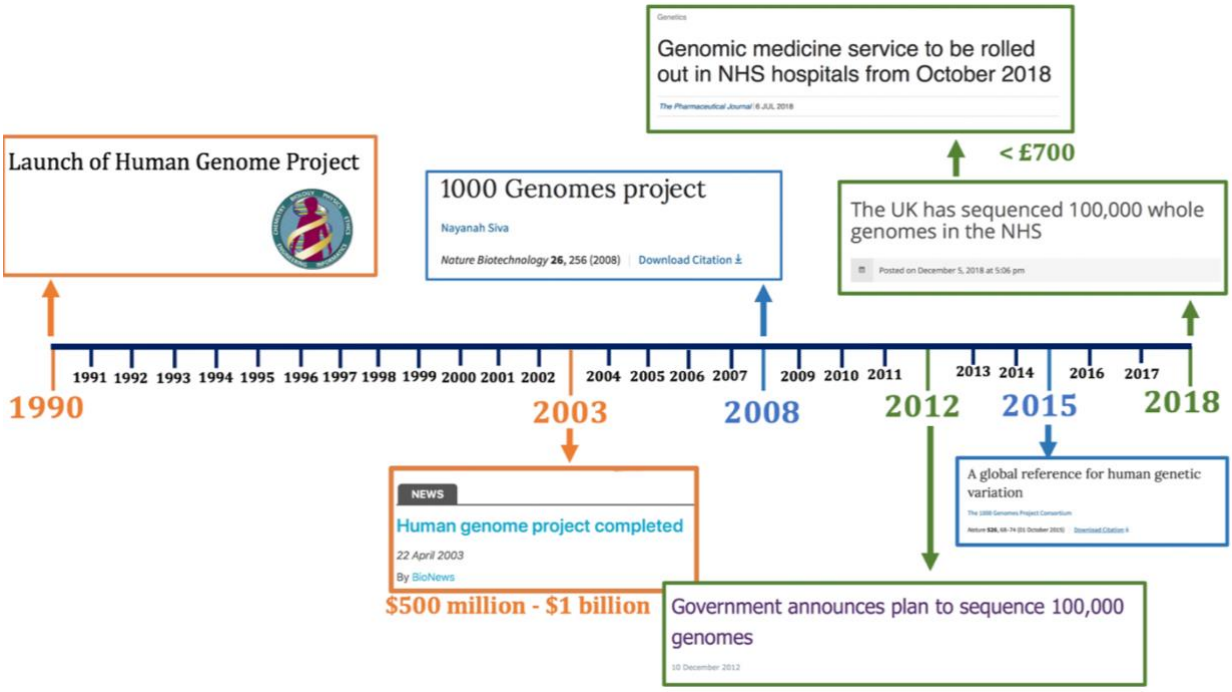


Figure 1 Timeline of genomic sequencing from 1990 to 2018.

1.5 The current state of genetics in healthcare

1.5.1 100,000 Genomes Project – what next?

Off the back of the successful 100,000 Genomes Project, the Government has announced an ambitious plan to sequence 5 million genomes by 2024 (Department of Health and Social Care, 2018). This will be a collaborative effort between the NHS, industry experts and research projects (the NHS and UK Biobank are expected to sequence 1 million genomes). To help reach this target, patients with a rare disease or certain types of cancer will continue to be offered WGS on the NHS, with the option to donate their data to research. This offer will also be made to seriously ill children. Further, the NHS considered offering healthy individuals the opportunity to pay for WGS (Photopoulos, 2019). Upon concerns that this would create a two-tier healthcare system, the Government have now announced plans to sequence the DNA of 5 million healthy volunteers for free (Sample, 2019).

Now, there are 3000+ researchers worldwide working on genomics, with the key aim to understand more about disease *before* it develops (Caulfield, 2019). Also, genomic medicine is being integrated into mainstream healthcare. This means that clinical specialists will have to order genetic tests and communicate the test results to patients. To make this transition easier, clinicians will use the National Genomic Test Directory to ascertain patients' eligibility for genetic tests, which includes WGS. Figure 2 illustrates a sample of this directory.

Clinical Indication	Target/Genes	Test Method Interim	Test Method Phase 1	Test Method Phase 2	Test Method Phase 3
Likely common aneuploidy	Genomewide	Common aneuploidy testing	Common aneuploidy testing	Common aneuploidy testing	Common aneuploidy testing
Common aneuploidy testing - prenatal	Genomewide	Common aneuploidy testing	Common aneuploidy testing	Common aneuploidy testing	Common aneuploidy testing
Possible structural chromosomal rearrangement - karyotype	As determined by indication	Karyotype	Karyotype	Karyotype	Karyotype
Possible structural or mosaic chromosomal abnormality - FISH	As determined by indication	FISH	FISH	FISH	FISH
Chromosomal mosaicism - karyotype	Genomewide	Karyotype	Karyotype	Karyotype	Karyotype
Chromosomal mosaicism - microarray	Genomewide	Microarray	Microarray	Microarray	Microarray
Acutely unwell children with a likely monogenic disorder	Paediatric disorders (486)	WES or Large Panel	WES or Large Panel	WGS	WGS
Acutely unwell children with a likely monogenic disorder	Genomewide	Microarray	Microarray	WGS	WGS
Ultra-rare and atypical monogenic disorders	Relevant panel(s) in PanelApp	WES or Large Panel	WGS	WGS	WGS
Ultra-rare and atypical monogenic disorders	Genomewide	Microarray	Microarray	WGS	WGS
Congenital malformation and dysmorphism syndromes - microarray and sequencing	Relevant panel(s) in PanelApp	WES or Large Panel	WGS	WGS	WGS
Congenital malformation and dysmorphism syndromes - microarray and sequencing	Genomewide	Microarray	Microarray	WGS	WGS
Congenital malformation and dysmorphism syndromes – microarray only	Genomewide	Microarray	Microarray	Microarray	Microarray
Intellectual disability – microarray, fragile X and sequencing	Intellectual disability (285)	WES or Large Panel	WGS	WGS	WGS
Intellectual disability – microarray, fragile X and sequencing	Genomewide	Microarray	Microarray	WGS	WGS
Intellectual disability – microarray, fragile X and sequencing	FMR1 STR	STR testing	STR testing	WGS	WGS
Intellectual disability – microarray only	Genomewide	Microarray	Microarray	Microarray	Microarray
Angelman syndrome	AS/PWS critical region	Methylation testing	Methylation testing	Methylation testing	Methylation testing
Angelman syndrome	AS/PWS critical region	MLPA or equivalent	MLPA or equivalent	MLPA or equivalent	MLPA or equivalent

Figure 2 Sample of the National Genomic Test Directory – a tool for establishing which genomic test a patient is eligible for.

1.5.2 What genetic tests do the NHS currently use?

The NHS already offers genetic testing in certain circumstances, summarised below.

1.5.2.1 High penetrance mutations

These are genetic mutations that are likely to be phenotypically expressed (Griffiths et al. 2005). For example, a mutation in one of the tumour-suppressing genes BRCA1 or BRCA2 can increase a woman's risk of developing breast cancer from the average of 12% to approximately 70% (National Cancer Institute, 2018). Patients are eligible for genetic testing if they have a family history of cancer and/or a relative with the mutation (NHS England, 2018b).

1.5.2.2 Monogenic disorders

These are disorders that derive from a single version of a gene. If a patient carries particular mutations of the gene, they will definitely develop the disease at some point. An example is Huntington's disease: those with a family history are eligible for genetic testing on the NHS (Huntington's Disease Association, 2019).

1.5.2.3 Cancer

The existence of particular mutations in a cancer can indicate the most effective treatment interventions for a patient (Dancey et al. 2012). Genomic testing to improve our understanding of a cancer's genetic base is fairly common and is used for a range of cancers, including brain, breast, and skin, as well as colon cancer and leukaemia (NHS England, 2016).

1.5.3 Genomics – a controversial topic

The unique arrangement of the NHS and its position as the world's largest integrated healthcare system currently enables vast numbers of patients to reap the benefits of genomic medicine (NHS England, 2018). However, genomic medicine opens up key ethical challenges as it is an approach to healthcare in which there is something of a grey area between research and clinical care: data obtained for research purposes may lead to findings of clinical significance, and patients can consent to the sharing of their genetic data for research purposes (Bertier, Cambon-Thomsen, & Yoly, 2018). Hallowell (2018) recognised that the ethical guidelines for both disciplines are distinct and, as of yet, no new ethical model has been implemented that

sees the convergence of the two domains. The question of whether patients have a responsibility to participate in research, and what this means for the consent process, has been debated.

Further, genomic data is unique because it can have implications for genetically related family members and can reveal unexpected findings. Consequently, WGS tests traditional ideas of consent and confidentiality. This is explored further below.

1.5.3.1 Confidentiality

Confidentiality is threatened by:

1.5.3.1.1 Genetic relatedness between individuals

Unlike other clinical tests, genetic tests can have implications for family members. Generally, confidentiality can be broken if there is an immediate threat to the life of another person; whether or not genetic test results represent such an immediate threat is debatable (Badzek et al. 2013). The complex ethical situation surrounding the legal responsibility of the doctor to provide a duty of care to one patient but also to maintain another patient's privacy is demonstrated in a recent legal case. In summary, a patient has brought a legal case against a hospital because they chose *not* to inform her about her father's Huntington's gene (Dyer, 2015). After her father's death, the patient discovered that she too carries the version of the gene for the hereditary disease and that her 8-year-old daughter has a 50% chance of carrying it. The patient states she would have had an abortion if she had known. This case will come to trial in November 2019 and may lead to changes in law regarding patient-doctor relationships.

A recent exploration of public opinion into this case indicated that a large majority of participants thought that, morally, the father should have told his daughter (Chapman et al. 2018). However, opinions were more polarized when asked about the daughter's right to the information and about whether the onus was on the NHS to disclose the genetic information. Indeed, 25% of participants believed that the NHS had no legal obligation to do so, whilst 35% of participants answered that the NHS 'absolutely' did. Overall, the authors concluded that such strong, polarised views indicated that the issues raised by the genetic relatedness between individuals are both complex and contentious.

1.5.3.1.2 Sharing clinical data

For genomic data to have clinical utility, it must be linked to patients' private medical records. This will help to unravel the complex relationship between genetics, what happens to us in our lives, and illness. This access requires the sharing of data across both national and international services. Indeed, plans to offer every patient the opportunity to participate in research and thus contribute to the National Genomic Data Resource demonstrates the need for the central collection of data (Hill, 2018).

1.5.3.2 Consent

WGS can reveal findings of diagnostic significance that are unconnected to the original reason for the genetic test (Mackley & Capps, 2017); some are sought out whilst others can be found unintentionally (Roche & Berg, 2015). Such 'additional findings' make the process of informed consent difficult.

The argument against informing patients of these findings is that, particularly if unactionable, i.e. no available medical intervention, they can cause psychological harm and distress (Ali-Khan et al. 2009) and may overload patients with information (Cho, 2008). However, if actionable, i.e. medical intervention is available, findings may inform treatment and improve patient choice (Caulfield et al. 2008). Indeed, a systematic review suggested these findings should be revealed if there is the possibility of effective treatment (Christenhusz, Devriendt & Dierickx, 2013). However, in a research setting without a duty of care, it is questionable whether we should be actively looking for these finding at all (Mackley & Capps, 2017). Further, it is very rare for researchers to return findings to patients as data is usually anonymised in research, so this must be consented to beforehand. For example, 100,000 Genomes Project participants had to opt in for the researchers to look for additional findings (Genomics England, 2018). These participants may receive these additional findings in several years time, as our knowledge of the human genome and the role of genes in disease expands, thus challenging the notion of 'informed' consent. This is complicated further by research that shows 54% of patients expect to be told about additional findings regardless of whether they had consented (Lucassen et al. 2019).

Further, young children are unable to provide informed consent regarding whether or not to have a genetic test, and yet the outcome has the potential to completely alter the course of their life (Selita, 2019). Similarly, *informed* consent is not possible without genetic knowledge or an understanding of all potential uses of their data. However, research shows poor levels of genetic knowledge even amongst the well-educated (Chapman et al. 2017) and organisations outside of the original institution have often been allowed third party access to data (Selita, 2019).

Overall, due to the range of possible findings (e.g. monogenic disorder versus carrier status), the implications for relatives, our limited but growing knowledge of the effects of individual genetic variants, as well as the difficulties surrounding the very notion of 'informed' consent, it is difficult to design a single consent protocol for 'additional findings' in healthcare (Mackley & Capps, 2017).

The next section will explore the extent of the public's understanding and knowledge of genetics.

1.6 Public knowledge & understanding of genetics

1.6.2 Summary of the literature

Studies indicate that research participants have relatively little knowledge of basic genetic concepts (Chapman et al. 2019; Haga et al. 2013; Lanie et al. 2004; Richards, 2016; Walter et al. 2004). Indeed, one study found a mere 1.2% of participants were able to correctly answer all 18 basic genetic literacy questions, and that those from the UK scored significantly lower than US participants (Chapman et al. 2019). Below is a summary of the main nuances and complexities of the public's understanding of genetics.

1.6.2.1 Heredity versus molecular genetics

The public display a better understanding of the genetic relatedness between family members (Kessler, Collier, & Halbert, 2007) and the fact that we inherit traits from our parents (Catz et al. 2005; Molster et al. 2009) than their understanding of the structure and/or function of genes. Other studies have also shown there is a greater understanding of heredity than there is of molecular genetics (Christensen et al. 2010; Haga et al. 2013; Lanie et al. 2004). For example,

in a survey of 300 participants aged 18-70, scores were considerably better on questions concerning the inheritance of disease than on questions on chromosomes and cells (Haga et al. 2013).

1.6.2.2 The role of personal experience

Personal experience of a health condition, e.g. a family member having a disease, can shape how an individual perceives its causal nature and hence their risk for it (Lucke et al. 2008; Walter et al. 2004). Indeed, Walter et al's (2004) meta-analysis explored the personalisation process of familial disease risk. The researchers found that an individual's perceived vulnerability to genetic disease was influenced by their relative's premature death or disability, their age at death and/or gender, and their emotional closeness to the relative.

More general personal experiences and beliefs also shape people's understanding of disease. For example, when unsure about the genetic relatedness of extended family members, people guess based on the strength of the social relationship (Richards, 2016), i.e. people with whom they are emotionally closer to are seen as more genetically related. Further, the public tend to choose causal variables of disease depending on their personal beliefs. For example, a study found that smokers were largely sceptical of a media story concerning the detection of a genetic variant linked to heightened nicotine addiction (Waters, Ball & Gehlert, 2017). The smokers' mistrust arose when their own beliefs about genetics clashed with the scientific evidence presented to them.

1.6.2.3 Physical versus behavioural traits

Research shows that the public believe that behavioural traits are more strongly associated with the environment, whereas physical traits are more strongly associated with genetics (Condit et al. 2004; Lanie et al. 2004). This is consistent with previous research that indicates perceptions of genetic attribution follow a certain pattern (Morin-Chassé, 2014; Parrott, Silk, & Condit, 2003; Shostak et al. 2009): the more biological a trait is, the bigger the perceived role of genetics in affecting that trait. Interestingly, people display the most confidence when estimating the genetic attribution of traits that are either highly associated with biology, e.g. height, or far from biology, e.g. being liberal or conservative (Morin-Chassé, 2014).

1.6.2.4 Gene-environment interaction & genetic determinism

The public perceive the role of both genetics and the environment to be important, however struggle to understand that these two factors can interact (Condit & Shen, 2011; Condit et al. 2009; French et al. 2000). French et al's (2000) study found participants regarded people who smoke or who have a family history of heart disease as very likely to experience a heart attack. However, when both risk factors were combined, participants did not increase their estimations, i.e. did not consider a gene-environment interaction.

Further, whilst some qualitative research indicates people have some implicit grasp of gene-environment interaction (Lanie et al. 2004), other qualitative research shows that once an individual decides a condition is behaviourally motivated, they consequently find it difficult to comprehend an alternative causal factor (Morris et al. 2003). Contrastingly, other research shows that most early adopters of personalised genomics exhibited a refined knowledge of the interaction between genes and environment (Gollust et al. 2012). However, the participants' status as early adopters of personalised genomics differentiates them from the average member of public who may not have even heard of 'personalised genomics'.

The inability to understand the interaction between genes and environment is tied in with 'genetic determinism' – the belief that a version of a gene for a trait will always lead to the development of that trait, rather than merely increasing the probability of its occurrence (Resnik & Vorhaus, 2006). Whilst some research shows the public display beliefs consistent with genetic determinism (Lanie et al. 2004; Parrott et al. 2012) other research found this belief to differ depending on the sample. For example, genetic determinism has been found more often in people who hold discriminatory beliefs, such as racism and sexism (Condit, 2011), or people who have less genetic knowledge (Chapman et al. 2019).

1.6.2.4.1 Impact of the media on beliefs of genetic determinism

Research into the impact of media reports of genetics shows an effect that is more subtle than an immediate endorsement of beliefs consistent with genetic determinism (Morin-Chassé, 2018). With reports of *medical* genetics, people accept a single, specific finding and adjust their genetic beliefs regarding that single, specific condition accordingly, e.g. breast cancer or diabetes (Smerecnik, 2010). However, with media reports of findings in *behavioural* genetics,

the public extrapolate the perceived importance of genetics to other, unmentioned traits, e.g. generalising a genetic finding related to violent behaviour to gambling addictions (Morin-Chassé, 2014). An increase in genetic determinism is harmful not only because it goes against scientific knowledge, but also because it can reinforce pre-existing intolerant attitudes (Gericke et al. 2017).

Regardless of the source of the public's misunderstandings, it is essential that we develop an understanding of the way in which peoples' knowledge of genetics affects their acceptance of genomic medicine. The following section will explore the public's attitudes towards genomics and demonstrate the importance of encouraging support for genomic medicine.

1.7 Public opinion of & attitudes towards genetic data

1.7.1 Review of the literature

A study in 1995 found that the general public views genetics as a 'double edged sword' (Michie et al. 1995, pg. 250). Indeed, due to the controversial nature of genomics (see section 1.5.3) this perspective has largely been maintained, with more recent research demonstrating the public still see genomic medicine as representing both great potential and great risk (Henneman et al. 2013; Wellcome Trust, 2016). Below is a summary of the literature regarding the public's main concerns and perceived benefits of genomic medicine.

1.7.1.1 Concern: commercialisation

One concern surrounding the use of genetic data in healthcare arises from the potential for these data to be used by commercial companies without obvious benefit to the public (Trinidad et al. 2010); 30.7% of participants in one study were concerned about their genetic data being used for marketing (Middleton et al. 2019). Another study found that 17% of participants would opt against commercial companies using their health data for any kind of research (Wellcome Trust, 2016). In the same study, 25% of participants indicated their concerns regarding data sharing outweighed the potential benefits from research, and 54% of participants indicated their highest priority was that the NHS sought permission for the commercialisation of their data - even if this meant that research would be halted. Similarly, Hapgood et al. (2004) found that the single most significant factor in deciding whether to

participate in UK Biobank was access to data; participants were content with the NHS and universities accessing their data but displayed dissatisfaction with access by third parties, e.g. pharmaceutical companies.

A potential source of this discontent could be the lack of public trust regarding commercial entities (Chalmers & Nicol, 2004; Ford et al. 2017; Levitt & Weldon, 2005). Indeed, one study found that for 94% of participants, their level of trust in an organisation would influence whether they would share their data with it (Open Data Institute, 2018). Another source of the dislike of commercial companies may be the belief that combining medical work with monetary benefit is ethically questionable (Haddow et al. 2007); some people believe that companies should not gain commercial benefit from patients' donations of their genomic data.

Further, whilst some research discovered that the public are usually more aware of commercial companies' role than is widely believed (Aitken, Cunningham-Burley & Pagliari, 2016), other studies show a lack of public understanding regarding the NHS's reasons for collaborating with commercial companies and how these companies use the data and play a part in healthcare (Castle-Clarke, 2018; Wellcome Trust, 2016). In fact, higher levels of acceptance of commercial companies has been linked to being more informed about how the data would be used (Jack & Womack, 2003; Wellcome Trust, 2016).

1.7.2.2 Concern: discrimination

A second common concern is the risk of discrimination and/or misuse of health data when it is used in research (Anderson, 2015). Despite many legal protections against it (Middleton, 2018), the fear of genetic discrimination is significant amongst the public (Wauters & Van Hoyweghen, 2016). Some research indicates that the concern that a dichotomous society could arise, with some people having 'good genes' and others having 'bad genes', has actually increased over the years (Henneman et al. 2013). In fact, public concerns that genetic discrimination could arise at a societal level in the form of a eugenics-like movement has been found in multiple studies (Fox, 2002; Geller, Bernhardt, & Holtzman, 2002; Gottweis, 2002; Vines, 1997).

On an individual level, participants have cited as a major concern firstly the potential for the NHS to prioritise other patients due to their genetics (Shickle, 1997; Wellcome Trust, 2013), and secondly the possibility that genetic results may lead to increases in insurance costs (Cook, 1999; Geer et al. 2001; Hall et al. 2005; Lemke et al. 2010; Stockdale, Cassell, & Ford, 2018) and decreases in employability status (Geller et al. 2002; Tambor et al. 2002). Indeed, in a study of 9 focus groups discussing the implications of genetic research, the topic of discrimination relating to employment and insurance came up in every group (Bates et al. 2005). This study also found that African Americans believe racial discrimination may arise from genetic research more than their white counterparts - a finding that has been replicated in other studies (Goldenberg et al. 2011; Peters, Rose, & Armstrong, 2004; Suther & Kiros, 2009; Middleton et al. 2018; Zimmerman et al. 2006).

1.7.2.3 Concern: data sharing & security

1.7.2.3.1 Access by research institutions

More generally, the public have also expressed fears about sharing their medical data for research purposes that includes sharing across different institutions and outside of their country of residence (Majumder, Cook-Deegan & McGuire, 2016). Only 24% of UK participants indicated they would trust medical research institutions with their data (Open Data Institute, 2018). Indeed, one study found that whilst 96% of participants supported the sharing of data for a patient's care, this decreased to 74% when the purpose was medical research (New Economics Foundation, 2010).

The public worry that research using their genetic data could damage their medical privacy (Anderlik & Rothstein, 2001; Gill & Richards, 1998; Kaufman et al. 2009), result in the copying and planting of their DNA at a crime scene, or the government and/or friends and family finding out something that the individual did not opt to disclose (Middleton, 2017).

1.7.2.3.2 Security of data in the NHS

However, it is not just research institutions that prompt anxiety from the public. Whilst trust in the NHS is high (Healthwatch England, 2018; Understanding Patient Data, 2018), research indicates the existence of a 'gulf' between how the public think the NHS could use their data and reality (New Economics Foundation, 2010). In fact, only 20% feel well-informed about

how their genomic data could be used (Healthwatch England, 2018), and one study found 71.3% of participants are doubtful that the NHS can assure full security of electronic health records (Stockdale, Cassell, & Ford, 2018).

1.7.1.4 Concern: genomic data is 'different'

A further barrier to genetic data sharing is 'genetic exceptionalism' – the perception that the genome is unique and distinct from other types of data, e.g. mental or sexual health data (Davies, 2017). Indeed, as discussed, there are some aspects of genetic data that make it different to other types of data. For example, genetic data challenges traditional ideas of consent and therefore people are concerned about implications for family members when having a genetic test themselves. However, there are some other concerns that are perhaps less realistic, such as the idea that our current knowledge of genetics is enough for it to be used in ways to edit characteristics of unborn babies, e.g. having a scheme in which parents can choose the intelligence of their unborn child. This could be linked to the misguided tendency to see genetic data as highly predictive and definitive, i.e. genetic determinism (Clayton, 2003; See section 1.6.2.4), or the related notion that genetic technology and using genetic data in healthcare is 'playing God' (Bates et al. 2005; Henneman, Timmermans, & Wal, 2006; Lassen & Jamison, 2006; Tambor et al. 2002). Regardless of its cause, 'genetic exceptionalism' can cause patients to have increased feelings of vulnerability and anxiety around sharing their genetic data which, whilst it is important for the public to have an awareness of the nature of genetic data, can lead to a high missed opportunity cost (Davies, 2017).

1.7.1.5 Are these concerns legitimate?

Concerns that are held by the public need to be examined and evaluated. The extent to which the concern is reasonable has different implications for genomic medicine. If a concern is more legitimate, we may have to consider changes to practice, but if the concern is more due to a misunderstanding, we may need to reassure the public through increased communication of the reality of the situation. However, it is important to recognise that whilst *legally* a concern may not be 'legitimate', there is always the possibility of accidental or illegal data access and/or leakage. Therefore, we can provide the public with the necessary information but should also ensure this does not lure them into a false sense of security, i.e. we should provide the information in an unbiased, accurate and balanced way.

For example, the most recent code on genetic testing and insurance states that insurers are only permitted to request a genetic test result if the test is both for Huntington's disease and for life insurance equalling above £500,000 per person (Government and the Association of British Insurers, 2018). This rule will be reviewed every 3 years and thus is subject to change. Presently, however, we could be providing the public with this information when they raise concerns about discrimination from insurers. Similarly, due to the current lack of genetic testing occurring in workplaces and the limited predictive value of genetic tests, specific legislation against genetic discrimination from employers has not yet been created (UK Parliament, 2009). Instead, employees are protected by broader laws such as the Disability Discrimination Act (1995), the Human Tissue Act (2004) and the Human Rights Act (1998) and the situation is monitored closely by the UK Government. This information could also be provided to the public when they raise concerns about genetic discrimination.

However, Selita (2019) noted that there have been some suggestions as to how to use genetic information in the selection of employees. For example, the US military have been recommended to look at the genetic profiles of potential military personnel to establish their genetic ability to tolerate stressful situations (The MITRE Corporation, 2010). Therefore, whilst current practice does not include the use of genetic information for employment purposes, the future is uncertain. Further, Selita (2019) recognised that the current trend of diminishing social provisions, coupled with ever-present health difficulties, means that an increasing portion of the public are turning to private insurance. Also, mental health illnesses are often undiagnosed (Mental Health Foundation, 2016); genetic information offers an instrumental tool to establish an individual's risk for health problems and consequently determine their insurance premium. Overall, the growing popularity of private insurance and the invaluable nature of genetic information to insurance companies arguably presents a case for changes to the rules surrounding access to genetic information.

Further, regarding worries about the security of data in the NHS, the public may be assured to know that genetic data, due to its sensitivity, falls under 'special category data' and thus gets extra protection (Information Commissioner's Office, 2019). For example, NHS staff are allocated 'smartcards' to access data. The amount of data they can access is automatically

adjusted based on their job, and they are only allowed access if they have good reason, e.g. to provide care (NHS England, 2011). Cyber security is recognised as an ongoing risk and thus managed at board level within the NHS - they have a Cyber Security Programme. Further, the NHS must store and secure genetic data in line with the rules of the Data Protection Act (NHS England, 2018c), which includes a legal obligation to apply suitable measures to protect such data (Data Protection Act, 2018), and the EU General Data Protection Regulation (GDPR). In fact, GDPR was the first legal framework to explicitly identify the category of genetic data (PHG Foundation, 2019).

However, having laws in place and a Government that oversees the situation by no means cancels out all serious concerns. There is still the possibility of negligence in data storage, accidental leaking of data, the illegal use of or access to information, or indeed the use of genetic information in specific circumstances, such as the recent use of a genealogy website to compare a suspect's DNA to the genetic profiles of the website's customers (Solon, 2018). Therefore, whilst we can increase communication of the laws and legislation surrounding genetic data, we should not use this information to ignore or diminish the public's concerns; there is a real possibility that some could still occur. All in all, the public should be provided with accurate and unbiased information regarding how data is stored and the way in which it is protected. Importantly, their decision as to whether or not to donate their genetic data is entirely their own and we should be using this information to inform, not manipulate, their choice.

With regards to concerns about commercial companies, if an individual gives consent to the use of their genetic data to research as well as for diagnostic testing within the NHS, this includes access by commercial partners. The commercial value of genomic data has the potential to bring large benefits to the NHS (UK Parliament, 2018) and collaborations with industry are required so that the NHS have sufficient funds to sequence, interpret, and store genetic data as well as develop new medicines (Life Sciences Industrial Strategy Board, 2017). For example, the NHS collaborated with the commercial technology provider Illumina during the successful 100,000 Genomes Project (Genomics England, 2016). Despite the necessity of commercial collaborations, this is a more legitimate concern because commercial companies are actually allowed access to people's genetic data. Perhaps changes to policy, such as a rule

whereby we must inform the public exactly which commercial organisations are allowed access, could be made.

1.7.1.6 Benefit: healthcare

Despite the aforementioned concerns and the lack of genetic knowledge shown in some studies (Chapman et al. 2019; Haga et al. 2013; Lanie et al. 2004; Richards, 2016; Walter et al. 2004) there is widespread public confidence that genomics will confer many benefits to healthcare (Hahn et al. 2010; Horrow et al. 2019; Health Research Authority, 2018; Wellcome Trust, 2016). As the perceived usefulness of genetic testing increases, so does the likelihood that it will receive support (Gaskell et al. 2000). One study found that 79.9% of participants cited improving their health as an important motivator to take part in genetic testing (Gollust et al. 2012). In fact, an Australian study found participants attributed higher importance to the maximisation of healthcare benefits from genomics than gaining specific consent for use of their genetic data in new studies (Critchley, Nicol, & McWhirter, 2017). The following section will explore areas within genomics that inspire public enthusiasm and excitement.

1.7.1.6.1 Impact on treatment

Research shows the public are optimistic about the possibility of developing both improved and new treatments as a result of genomic medicine. For example, public dialogue workshops found the majority of participants were excited about the possibility of better NHS treatments (Health Research Authority, 2018), and one of the most cited benefits in another study was the possibility of new treatments (Hahn et al. 2010). Indeed, 44% of participants who had originally opposed commercial companies having access to genetic data would reverse that decision if it meant the development of novel treatments would not take place (Wellcome Trust, 2016). Further, one study found that 90.7% of participants displayed confidence that genomics will help doctors choose the best treatment for patients (Horrow et al. 2019)

1.7.1.6.2 Diagnoses

Studies also show support for genomic medicine because it could help patients get an accurate diagnosis. For example, a study exploring opinion of technological interventions found that 84% of participants supported genetic screening tests for cystic fibrosis - the most supported intervention across the entire study (Calnan, Montaner, & Horne 2005). Other research

discovered that 91% of participants endorsed a statement that indicated a high likelihood that genomic medicine will help doctors diagnose rare diseases (Horrow et al. 2019).

1.7.1.6.3 Knowledge of genetic risk & prevention

The public also hold high hopes that genomic medicine will encourage preventative measures by providing information about genetic risk. Indeed, one study found that those who think that knowing the genetics of disease will encourage healthier lifestyles in those who are at genetic risk are less likely to be opposed to genomic medicine (Henneman, Timmermans, & Wal, 2006). The same study found that, whilst most participants originally wanted to remain in the dark about their genetic disease risk, once the possibility that the disease in question was preventable, interest increased to over 50% of participants. Similarly, another study found 78.4% of participants were motivated to take part in genetic testing because they wanted to know which diseases they were at risk for (Gollust et al. 2012), and qualitative research has found that attitudes to genomic medicine are heavily informed by the perceived utility of the genomic information (Nicholls et al. 2013).

However, whilst some studies indicate that genetic test results improve and/or increase users' healthy behaviours (Gordon et al. 2012; Kaufman et al. 2012; Roberts, Christensen & Green, 2011) other studies show that users of genetic tests make no changes to their behaviour (Bloss et al. 2010; Bloss et al. 2013; Hollands et al. 2016). Research exploring the impact of genetic results on anxiety also shows mixed findings. Some researchers suggest that genetic results may decrease anxiety in users (Hilgart, Coles, & Iredale, 2012), whilst others suggest results have no effect on anxiety (Bloss et al. 2010; McBride, Wade & Kaphingst, 2010; Roberts, Christensen & Green, 2011) or may increase anxiety (Samuel, Jordens, & Kerridge, 2010). Given these mixed findings, the perceived importance of the utility of genetic tests (Nicholls et al. 2013) is brought into question.

1.7.1.7 Benefit: the greater good

1.7.1.7.1 Societal benefit

More broadly, a key condition for public acceptance of sharing patient data and using it beyond individual care is that it will confer some benefit to society (Asthma UK, 2018; Wellcome Trust, 2016; Tully et al. 2018; Understanding Patient Data, 2018). Indeed, qualitative work found the majority of participants comment on the cruciality of genetic research to offer some societal

benefit (Stolt et al. 2002). In addition, the public appear keen to participate in research, with 80% of participants stating they would want their clinician to inform them about medical research they could get involved in (Ipsos MORI, 2011). In fact, a systematic review found that a main component of a trustworthy, supported organisation was the public's perception that they had the 'right motivations', i.e. public benefit (Stockdale, Cassell, & Ford, 2018). Research shows there may be an element of social organisation to individual decision-making: those who take part in medical research do so to demonstrate their connection with others and to confer a benefit to the public good (Dixon-Woods & Tarrant, 2009).

1.7.1.7.2 Scientific advancement

Finally, qualitative research indicates that agreeing to share genomic data is encouraged by the idea that it may benefit scientific and medical research (Thiebes, Lyytinen & Sunyaev, 2017). In support of this, 94.3% of participants trust that genomic medicine will help scientists discover facts about genes (Gollust et al. 2012) and 94.6% of participants believe it will help to answer difficult questions about disease in the next 5 years (Horrow et al. 2019). Overall, it is clear that the 'double-edged sword' description of genomics (Michie et al. 1995) still stands today. Now, an interesting and important avenue to explore is the factors that feed into public backing of genomics. To this end, the following section will explore the role of genetic knowledge.

1.8 Does knowledge of genetics link to acceptance of genomic medicine?

1.8.1 Deficit model – in support of a positive relationship between knowledge & acceptance

The assumption that a lack of acceptance of genomics is a result of a misunderstanding of genetics is based on the 'deficit model', that assumes scepticism and fear towards science and technology is due to a deficiency in public scientific understanding (Sturgis & Allum, 2004). There are scientific studies that provide support for this model (Hayes & Tariq, 2000; Morren et al. 2007). For example, one study found that one of the greatest predictors of the public's perceptions of benefits from biotechnology, which included genetic testing, was the degree to which an individual was informed about biotechnology (Pardo, Midden, & Miller, 2002).

With regards to genetics more specifically, a large UK study by the Human Genetics Commission (2001) found participants with low levels of genetic knowledge were least likely to acknowledge that developments in genetics may bring cures for disease. These participants were also the most likely to perceive genetic research as interfering with nature and thus as unethical. Further, an analysis of survey results from UK adults found those with higher scientific knowledge displayed more optimism about medical genetics and more enthusiasm about genetic testing (Allum et al. 2014) and, similarly, a study by Chapman et al. (2019) found a weak but positive relationship between participants' knowledge of genetics and their readiness to take a genetic test for medical reasons. A recent experimental study, which explored the effect of an in-class lecture in clinical genetics on undergraduates' attitudes towards genetic testing, also provided support for the deficit model (McClintock, 2019). The lecture included information on the limitations of clinical genetic testing, the types and classifications of genetic tests, and the benefits of prenatal testing. Participants who were exposed to the lecture displayed increased positive attitudes towards genetic testing compared to those who did not receive the lecture.

1.8.2 Evidence against a relationship between knowledge and acceptance

1.8.2.1 No evidence for a relationship

However, contradictory to the deficit model and the above studies, there is evidence of a null relationship between scientific knowledge and positive attitudes towards science. For example, Gottweis (2002) suggests that low levels of acceptance of gene therapy are caused not by a lack of knowledge but a lack of trust in scientists and technology. Further, an analysis of 1,308 survey responses from Dutch participants found genetic knowledge was not significantly associated with the participants' status of being either an 'opponent' or a 'supporter' of genetic tests (Henneman, Timmermans, & Wal, 2006). Rather, familiarity with a genetic disease was associated with a decreased likelihood of being opposed to genetic testing. Additionally, contrary to McClintock's (2019) experiment, another experimental UK study found the provision of a 10-minute scientific documentary about genomics had no impact on participants' overall attitude to genetic science and databanks (Sturgis, Brunton-Smith, & Fife-Schaw, 2010).

1.8.2.2 Evidence for a negative relationship

Finally, there is also some evidence of a *negative* relationship - one study discovered individuals who were most knowledgeable about genes were also the most concerned that genetic tests will not improve people's quality of life and may lead to eugenics, whilst those least knowledgeable were most unsure of how they felt (Jallinjoa & Aro, 2000). Further, in morally controversial fields of research, which arguably includes genetics, more-knowledgeable individuals have been found to hold stronger opposing views to research than those who were less informed (Evans & Durant, 1995).

1.9 The present study

Overall, the relationship between knowledge and attitudes is not well-defined. The present study aimed to investigate the hypothesis that fear towards genomic medicine is due to a lack of knowledge. The study explored genetic knowledge in three different ways: 1) participants' biological knowledge of genetics, 2) their clinical knowledge of genomic medicine, and 3) their perception of their genetic knowledge.

In addition, given the controversial nature of genomic medicine, I conducted two focus groups to explore public concerns, expectations, and opinion of the use of genetics in healthcare. To the best of my knowledge, this was one of the first focus groups gathering public opinion about the most recent NHS plans for genomic medicine. This was an exploratory study with no specific hypotheses.

1.9.1 Why the UK public?

1.9.1.1 Importance of research into the UK

Differing opinions on the topic of genetics may arise as a result of cultural sensitivities, historical influences (Gaskell et al., 1999) or country of residence (Gaskell et al. 2000), e.g. UK participants have shown greater support for genomic research than their Australian and American counterparts (Middleton et al. 2018). Therefore, the participants in my study may hold views that are not present within other cohorts. Further, it is important to specifically explore the views of the UK public given that the UK is the first country in the world to bring genomics into a mainstream healthcare system (Davies, 2017) and, in addition, has a healthcare system that differs vastly from other countries such as the US, Japan and Australia

(Duncan & Jowit, 2018). The fast pace of genomics in the UK may foster thoughts and opinions that are exclusive to those who reside in this country.

1.9.1.2 Importance of research into the public

The development of genome policies using only the opinion of experts limits the possibility of success as they would be unlikely to generate satisfaction in all stakeholders (Haga & Willard, 2006). Also, attempts to understand public attitude towards genomics could uncover sources of scepticism and gaps in knowledge, that in turn could be used to focus communication efforts (Henneman et al. 2013). On a more practical level, biobanks depend upon members of the public to donate samples and information (Samuel & Farside, 2018) and members of the public pay for research and are prospective users of the resulting products (Bates et al. 2005). Overall, the controversial nature of genetics means the involvement of the public is crucial to increase accountability in the decision-making process (Samuel & Farsides, 2018).

1.9.2 How to address the research question? Use of quantitative & qualitative methods

This project used a mixed methods design. Use of a quantitative design allowed for a reliable, generalisable and powerful conclusion. Contrastingly, whilst the qualitative results are not generalisable to the larger UK population, they allow for a deeper insight and may uncover complexities that had not been considered previously. A mixed-methods approach can enable the collection of different and sometimes opposing views (Greene, 2007). The combination of both studies' strengths is vital if we are to inform changes to practice based upon a full understanding of public perception of genetics in healthcare.

1.10 Chapter summary

In this chapter, I have outlined the current state of genomic medicine, as well as its history and hopes for the future. I discussed the controversial nature of genetic data, the public's concerns and expectations, and gave an overview of the evidence for and against the deficit model. This led us to one of my research questions: does knowledge of genetics predict acceptance of genomic medicine? The importance of research into public opinion directed us to my second

research question: what concerns and expectations do the UK public hold about genomic medicine? I concluded this chapter with a justification for my choice of a mixed methods approach.

Chapter 2: Quantitative study – Does knowledge of genetics predict acceptance of genomic medicine?

2.1 Introduction

Genomic medicine is becoming increasingly relevant and thus important for all members of the UK public. As the Chief Medical Officer acknowledged, the success of genomic medicine depends on the establishment of public trust (Davies, 2017). The question remains, however, as to the best methods of communication to achieve this aim. It is unclear whether attempts to improve genomic literacy and understanding will improve public acceptance of genomic medicine. Understanding and characterising the relationship between knowledge and acceptance of genomic medicine will help to guide educational efforts and resources. In turn, this will maximise the probability that the UK public will engage in and be supportive of the use of genetic data in healthcare.

On the one hand, some research indicates that such an educational undertaking will improve public acceptance. The deficit model suggests that the endorsement of a scientific belief or domain is intrinsically linked to how much scientific knowledge one holds (Sturgis & Allum, 2004). In other words, the more science you know, the more comfortable you feel with a particular scientific area. As mentioned in 1.8.2, there is literature to support this claim within the domain of genetics (Allum et al. 2014; Human Genetics Consortium, 2001; Chapman et al. 2019; Pardo, Midden, & Miller, 2002).

On the other hand, other research indicates that attempts to increase public understanding of genetics may have no impact on public acceptance (Henneman, Timmermans, & Wal, 2006; Sturgis, Brunton-Smith, & Fife-Schaw, 2010) or indeed may have the *opposite* intended effect (Jallinoja & Aro, 2002). These studies suggest that other variables are far more important in determining an individual's level of acceptance towards genetics, and thus resources may be

better spent elsewhere. These other variables include the level of trust in scientists (Gottweis, 2002), familiarity with a genetic disease and belief in the personal benefits of genetic testing (Henneman, Timmermans, & Wal, 2006), and how controversial the scientific application is (Bak, 2001).

Overall, we are left with scientific evidence to both support and counter the deficit model, and the nature of the relationship between genetic knowledge and acceptance of genomic medicine remains uncertain. Importantly, however, a review of studies between 1993 and 2008 into the deficit model found that the strength of the relationship depended on the specific *type* of scientific knowledge studied (Allum et al. 2008). For example, the relationship between general scientific knowledge and positive attitudes to genetically modified (GM) food was almost non-existent. However, *biological/genetic* knowledge was a strong predictor of positive attitudes to GM foods. Crucially, the overall relationship between biological/genetic knowledge and positive attitudes to genetic medicine was non-significant. Allum et al. (2008) concluded that future research should focus on specific aspects of scientific knowledge, rather than using a more general measure of scientific knowledge. The field of genetics has changed vastly since Allum et al's study in 2008 (see Figure 1). Therefore, my research is crucial if we are to identify the relationship between knowledge and acceptance as it exists within the UK public today.

Indeed, whilst some aforementioned studies specifically explored genetics (Allum et al. 2014; Chapman et al. 2019; Human Genetics Consortium, 2001; Jallinjoa & Aro, 2000; Sturgis, Brunton-Smith, & Fife-Schaw, 2010), other research has explored a multitude of domains. This includes public opinion on health data (Asthma UK, 2018; Wellcome Trust, 2016), personal data (Open Data Institute, 2018), medical data (Majumder, Cook-Deegan & McGuire, 2016) electronic health records (Stockdale Cassell, & Ford, 2018), biotechnology (Pardo et al. 2002) and gene therapy (Gottweis, 2002). Thus, there is a need for more research that specifically explores *genetic* knowledge and how that links with acceptance of the use of *genetic* data in healthcare.

The present study sought to specifically test participants' biological knowledge of genetics, defined by their knowledge of genes and how they work in the body, their clinical knowledge of genetics in healthcare, defined by their awareness of the current situation regarding the genomic medicine service in the UK, and their perception of their genetic knowledge. I also

measured participants' level of acceptance of the use of genetic data in UK healthcare. This included questions that were only relevant to the state of genomics in the UK (see 2.2.1.6). I aimed to test the hypothesis that fear towards genomic medicine is due to a lack of understanding of biological and/or clinical knowledge of genetics. I added perception of genetic knowledge as an exploratory measure. This study is novel because, to my knowledge, no previous studies have made this distinction between biological and clinical knowledge in order to explore their relationship with public acceptance of genomic medicine. Further, this study specifically looked at the UK public. Whilst some previous research into public knowledge of and acceptability towards genetics has been conducted in the UK (Calnan, Montaner & Horne, 2005; Castle-Clarke, 2018; Healthwatch England, 2018; Middleton, 2018), a large amount of research is focused in the US (Christensen et al. 2010; Condit et al. 2004; Kaufman et al. 2009; Kessler, Collier, & Halbert, 2007; Majumder, Cook-Deegan, McGuire, 2016; McClintock, 2019) or in Europe more broadly (Gaskell et al. 2001; Henneman et al. 2013; Pardo, Midden & Miller, 2002). Given the unique position of the UK as the worldwide leader of genomic medicine, the exploration of the relationship between knowledge and acceptance in members of the UK public is particularly pressing.

2.2 Method

This study was approved by the University of Bristol School of Psychological Science Research Ethics Committee on the 12th October 2018 (ethical approval code: 75841; see Appendix 1). The overall process of developing the survey included the following key steps: development, distribution, analysis of the first wave data and comparison with UK census data, targeted re-distribution, and final statistical analysis. The methodological

steps in this study have been summarised in Figure 3 below. The following sections will elaborate upon each step in this overall process.

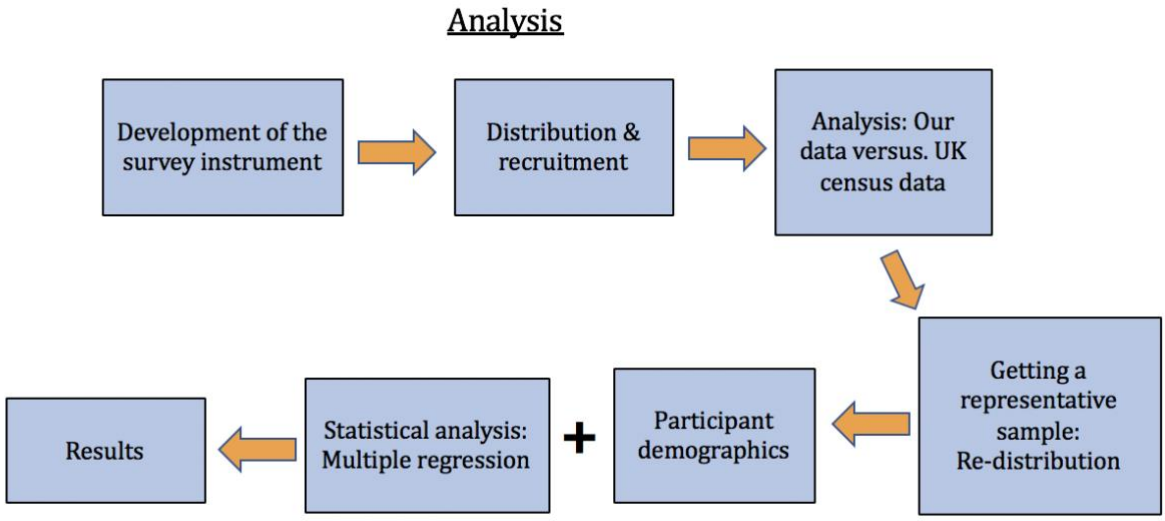


Figure 3 Overall steps of the development of the survey exploring people's biological knowledge of genetics, their clinical knowledge of genetics in healthcare, their perception of their knowledge, and their acceptance of genomic medicine.

2.2.1 Development of the survey instrument

The survey consisted of 6 sections: (1) Demographic information; (2) Experience with genetic testing, (3) Biological knowledge of genetics; (4) Clinical knowledge of the use of genetics in healthcare; (5) Perception of genetic knowledge; (6) Acceptance of the use of genetic data in healthcare.

The final instrument included a mix of newly constructed questions, questions that had been developed upon discussion with a genetic counsellor, and questions adapted from previous instruments. These instruments included: the Wellcome Trust Monitors (2012; 2016); the Public Understanding and Attitudes towards Genetics and Genomics survey (PUGGS; Carver et al. 2017); the International Genetic Literacy and Attitudes Survey (iGLAS; Chapman et al. 2017); questions from a Genes and Behaviour course assessment used at the University of Bristol (Haworth, 2018); and Your DNA, Your Say, a survey designed by The Society for Ethics Research Group at the Wellcome Genome Campus (Middleton, 2017). I chose questions from

these sources to test the four core topics: biological knowledge, clinical knowledge, perception of knowledge, and acceptance of genomic medicine. For further details regarding the exact source of each question, refer to Appendix 2.

2.2.1.1 Demographic information

We asked participants for general demographic information, i.e. age, gender, ethnicity, education, as well as information specifically relevant for this study, such as if participants worked for the NHS, when they were last taught about genetics, whether they come across genetics in the workplace, and finally whether they have any children. I added these additional demographic questions as I acknowledged that these factors may influence participants' knowledge of genetics and/or acceptance of genetics in healthcare and wanted to be able to control for them.

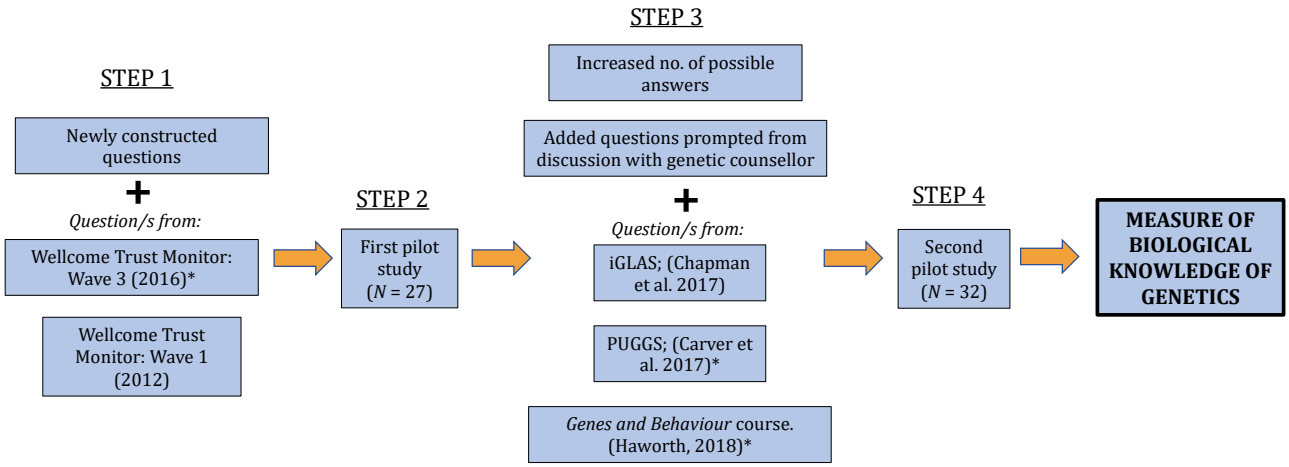
2.2.1.2 Experience with genetic testing

In addition to the above, I recognised that an additional confounding variable could be participants' experience with genetic testing. Indeed, research indicates a disconnect between the general public's and patients' perceived benefits of genomic medicine (Critchley, Nicol, & McWhirter, 2017). Thus, participants were asked questions such as (for all questions, see Appendix 2):

Have you or anyone close to you ever had a genetic test done by a doctor?

2.2.1.3 Measuring biological knowledge of genetics

We wanted to measure participants' level of knowledge regarding the biology of genes and the mechanisms of genetic influences. The overall process of the development of this measure is summarised in Figure 4. The following sections will elaborate on this process.



** = Some/all question/s adapted format and/or content adapted for the current survey.*

Figure 4 Steps in the development process of the biological knowledge of genetics measure.

2.2.1.3.1 Step 1: Initial instrument

An initial assessment was developed that included questions from the 2012 and 2016 Wellcome Trust Monitors and newly constructed questions. For example, I used a true/false question from the Wellcome Trust Monitor (2016) that tested participants' knowledge of the genetic influences from biological parents:

It is the mother's genes that determine the sex of the child

A question from the Wellcome Trust Monitor (2012) paper tested participant's knowledge of genetic probability:

A doctor tells a couple that they've got a one in four chance of having a child with genetic disease...

- *...if their first three children are healthy, the fourth will have the illness*
- *...if their first child has the illness, the next three will not*
- *...each of the couple's children will have the same risk of suffering from the illness*
- *...if they have only three children, none will have the illness*

The above question also motivated the development of two newly constructed questions, both of which presented a scenario in a similar way. For example, to test participants' knowledge of

the relative influences of genetics and the environment, I developed, piloted, and included the following question:

A patient is told that the results of their genetic test indicate they are at increased risk of a particular disease. Consequently, they decide to engage in healthier behaviours, such as exercising regularly and eating a more balanced diet.

- *…their increase in healthier behaviour will have no effect on their likelihood of developing the disease, because their genetic test indicates they will definitely develop the disease at some point*
- *…their increase in healthier behaviour will definitely decrease their likelihood of developing the disease, because the effect of the environment will always override the influence of their genes*
- *…their increase in healthier behaviour may lead to a decrease in their likelihood of developing the disease, because the way their genes work can be altered by their environment*

Additional newly constructed questions were developed through an exploration of common misunderstandings of genetics (Christensen et al. 2010; Dar-Nimrod & Heine, 2011; Klitzman, 2010). For example, inclusion of the false statement, '*Genes are always more important for how you look than how you behave'*, in the question *'Which statement is TRUE'*, tested participants' level of genetically deterministic beliefs and how they distinguish between physical and behavioural traits.

2.2.1.3.2 Step 2: First pilot study

We piloted the initial version of this measure and recruited 26 participants via email and social media (Female =15, Mean age =32yrs). Results indicated that overall scores displayed a ceiling effect and were skewed towards the 100% correct mark. For further details, see Appendix 3.1.

2.2.1.3.3 Step 3: Adjustment & addition of questions

The findings from the pilot indicated that, in order to eliminate the ceiling effect and the skewed nature of the data, I needed to increase the difficulty of the questions. Indeed, a large majority of the questions gave only two options for the answer (*True* or *False*), meaning that participants had a 50% chance of guessing the correct answer. Therefore, I rearranged the structure of the questions in order to generate 2+ possible answers. For example:

Which of the following is TRUE:

- *It is the mother's genes that determine the sex of the child*
- *If there is no family history of disease, then there is no chance of a newborn baby having said disease*
- *Humans share approximately 99% of their DNA with each other*

To increase the difficulty further, I incorporated questions adapted from multiple additional sources. These included: Public Understanding and Attitudes towards Genetics and Genomics survey (PUGGS; Carver et al. 2017); University of Bristol Genes and Behaviour course assessment (Haworth, 2018); and the International Genetic Literacy and Attitudes Survey (iGLAS; Chapman et al. 2017). The following is an example of a question from iGLAS (Chapman et al. 2017), which tests participants' knowledge of the role genes in the human body:

What is the main function of all genes?

- *Storing information for protein synthesis*
- *To provide energy to the cell*
- *To clear out waste from the cell*
- *To repair damage to the cell*

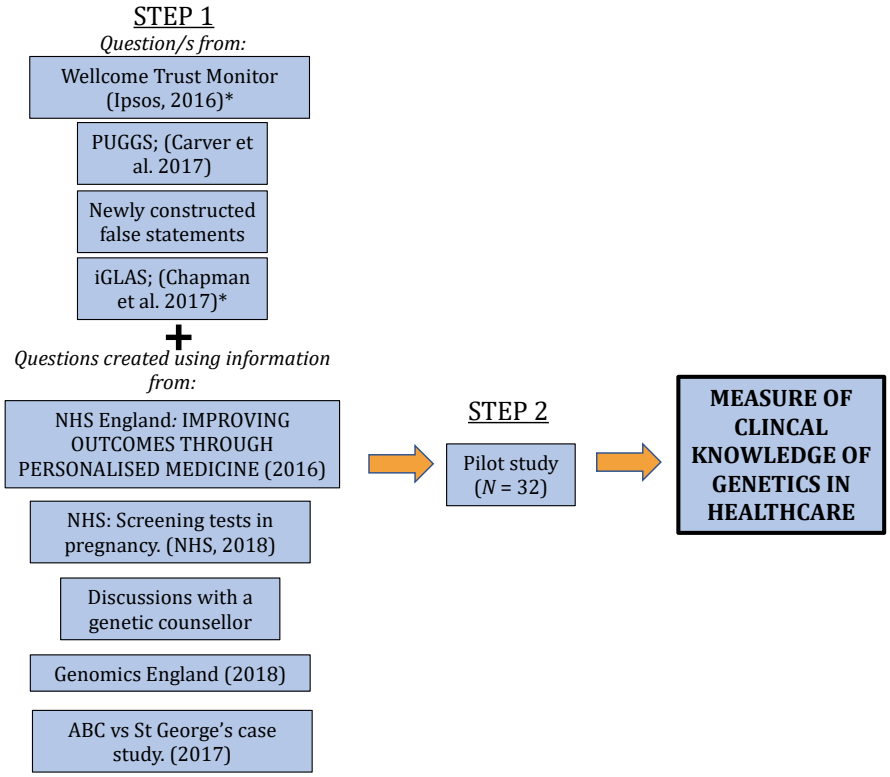
Finally, through my collaboration with the NHS West of England Genomic Medicine Centre, I organised a meeting with a genetic counsellor. We discussed the most common unknowns in the biology of genetics and their influence on healthcare, and two true statements were consequently incorporated into questions: '*Your genetic make-up can influence the way that you respond to medical interventions*' and '*Cancer can be caused by both inherited genetic mutations or mutations that are acquired throughout the lifetime'.*

2.2.1.3.4 Step 4: Second pilot study

A second pilot study recruited 32 participants through social media and email (Female =25, Mean age =34yrs). Results indicated a normal distribution of overall scores with an acceptable level of skewness (see Appendix 3.2 for results). Therefore, the revised version of this section was considered suitable for testing.

2.2.1.4 Measuring clinical knowledge of the use of genetic data in healthcare

The second section of the survey was designed to assess participants' knowledge of the current situation regarding the use of genetics in healthcare, i.e. their clinical knowledge. Figure 5 below indicates the overall process of this measure's development, and the following sections discuss each step in more detail.



** = Some/all question/s format and/or content adapted for the current survey.*

Figure 5 Steps in the development process of the clinical knowledge of genetics measure.

2.2.1.5.1 Step 1: Instrument development

Newly constructed (false) statements were created by the lead researcher and based upon common misunderstandings and misconceptions of genetics in healthcare. For example, research indicates the public hold unrealistic and exaggerated expectations of genomics (Evans et al. 2011). Thus, the following false statements were developed to test such inflated beliefs:

Using an individuals' genetic code, we are currently able to predict whether an individual will develop every single disease known to the medical community

Currently, the NHS has every individual patient's genetic code stored on their database

Other true/false questions were formulated through examination of the information on the websites of Genomics England (2018) and NHS England: Improving Outcomes Through Personalised Medicine (2016) and NHS: Screening tests in pregnancy (2018c). For example:

Through a number of genetic testing techniques, it is possible to detect genetic abnormalities in an unborn child

In addition, the recent 'Patient ABC versus St George's Healthcare Trust' case (Dyer, 2015), which challenges conventional ideas of medical confidentiality, motivated a question regarding legal responsibility and patient privacy. The question developed was as follows:

*It is against the law for a doctor to **not** disclose a patient's genetic test results to their close relatives as, given their genetic relatedness, the results may also concern them*

Another true/false question was added after discussions with a genetic counsellor revealed that many people are unaware of the exact role of a genetic counsellor:

A patient will only speak with a genetic counsellor if their genetic test result indicates they have a genetic predisposition to a disease

Finally, inclusion of a question adapted from the Wellcome Trust Monitor (2016), *(How much have you read or heard about genetic tests that predict the likelihood that a person will develop certain genetically influenced diseases or conditions, such as heart disease, cancer and Alzheimer's?),* motivated yes/no questions that explored participants' self-reported awareness of specific phrases and projects. For example:

Have you heard of the 100,000 Genomes Project?

Have you heard of the phrase 'personalised medicine?'

2.2.1.5.2 Step 2: Piloting

This version of the measure was piloted, and participants recruited via social media and email (Female =25, Mean age =34yrs). Results revealed a normal distribution of overall scores with an acceptable level of skewness (see Appendix 3.3 for further detail). Thus, the measure was deemed acceptable to include in the final survey.

2.2.1.5 Measuring participants' perception of genetic knowledge

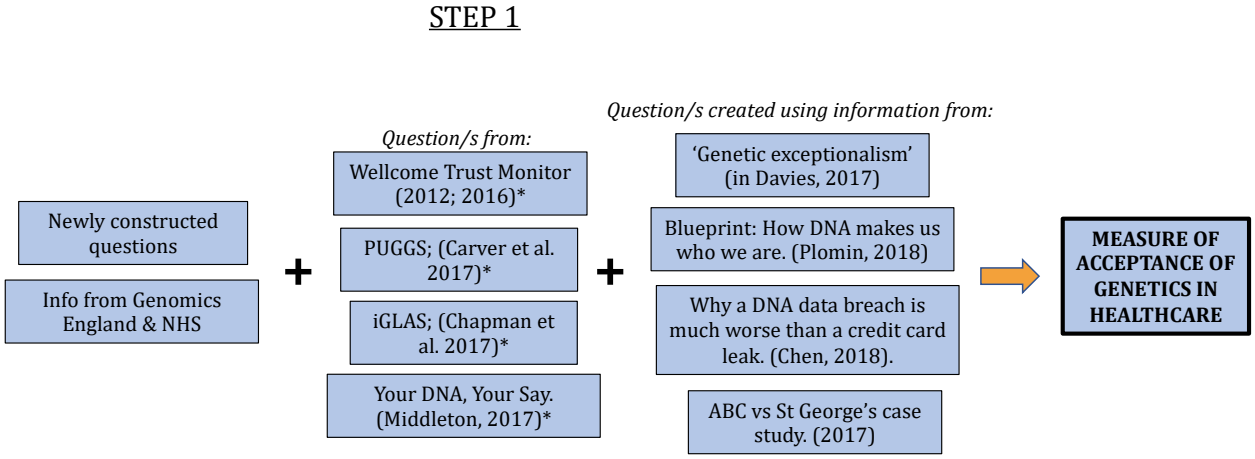
In addition to testing participants' biological knowledge of genetics, I also included questions to measure participants' *perception* of their genetic knowledge. This was an exploratory measure, included in order to examine whether the public accurately rate their knowledge of genetics, and included a total of 4 questions. For example, I used a question from the Wellcome Trust Monitor (2012) to explore participants self-reported understanding of the ethical issues within genetic research, from 'Very good' to 'Have not heard the term'. Another question was also from the Wellcome Trust Monitor (2012):

When you hear the term DNA, how would you rate your understanding of what the term means?

In addition, I incorporated similar questions from the 2016 and 2013 version of the Wellcome Trust Monitors that asked participants to rate their understanding of the terms 'genetically modified' and 'human genome', respectively.

2.2.1.6 Measuring acceptance of genetics in healthcare

The final measure in the survey was designed to explore how accepting participants feel towards the use of genetic data in healthcare. Figure 6 below summarises the development process, and the following sections will elaborate upon each step of the process.



** = Some/all question/s format and/or content adapted for the current survey*

Figure 6 Process of the development of measure of acceptance of genomic medicine.

First, participants were given a brief overview of the current situation regarding the use of genetic data in healthcare in the UK, adapted from information from the websites of NHS England (2018a) and Genomics England (2019). This was to ensure that participants answered questions on an informed basis. For example, '*Overall, the aim is to create a new genetic medicine service for the NHS … patients may be offered a diagnosis where there wasn't one before or may be able to make more informed decisions regarding their treatment.'*

Next, questions were adapted from the Wellcome Trust Monitors (2012; 2016); iGLAS (Chapman et al. 2017); PUGGS (Carver et al. 2017); and Your DNA, Your Say (Middleton, 2017). For example, to test participants' trust in the NHS, the following question was adapted from Chapman et al. (2017) and measured on a 6-point scale from 'Strongly agree' to 'Strongly disagree':

I do not trust the healthcare system in the UK because it might misuse genetic data obtained from patients

Additional questions were incorporated following research into common concerns about genetics in healthcare (Haddow et al. 2007; Nowlan, 2002; Zaidi, 2018). For example, a news article entitled 'Why a DNA data breach is much worse than a credit card leak' (Chen, 2018) motivated the development of the following question, again to explore whether the present participants hold beliefs that are consistent with genetic exceptionalism (Davies, 2017):

I would be more concerned about my genetic data being leaked than my credit card information

Finally, a question was developed following attendance at Professor Robert Plomin's annual lecture at the University of Bristol - Blueprint: How DNA Makes Us Who We Are (Plomin, 2018). In this lecture, Plomin discussed the benefits of knowing your genetic information, and the notion that 'knowledge is power':

Finally, imagine yourself in the following scenario. You have an opportunity to have a genetic test that may reveal that you are going to develop a condition that currently does not have any medical treatment options. Which statement do you agree with most?

- *Knowledge is power: I would rather have the genetic test, so that I can prepare financially, emotionally, and practically if I am found to have a genetic variant that will lead to an (currently) incurable disease*
- *Ignorance is bliss: I would rather not have a genetic test, because if I am found to have a genetic variant that will lead to an (currently) incurable disease I would worry about it too much to be able to carry on enjoying my life to the full*
- *I'm not sure*

We did not pilot the acceptance measure as I expected a range of opinions and there were no right or wrong answers. Therefore, it was unlikely to result in a ceiling (or floor) effect in the same way as the knowledge measures of the survey.

2.2.1.7 Additional questions

Six questions did not explicitly explore acceptance of genomic medicine but were included to form part of an additional exploratory analysis of public attitude towards genomic medicine. For example, motivated by the aforementioned legal case (Dyer, 2015), the following question was designed to explore participants' attitudes towards the level of responsibility of genetically related family members and again measured on a 6-point scale of agreement:

Family members share many genetic traits and may have the same genetic abnormalities associated with disease. Therefore, all immediate family members must give consent before an individual in that family gets a genetic test for medical reasons

To investigate participants' levels of genetic determinism, I asked participants to indicate their agreement with a statement from Chapman et al. (2017):

I believe that my destiny is written in my genes

Further, to explore participants' attitudes towards genomics' approach to medicine, I asked participants to rate their agreement with another statement from Chapman et al. (2017):

Preventing health problems is preferable to curing health problems

Finally, I included a set of questions to explore the concept of genetic exceptionalism (Davies, 2017; see section 1.7.2.4), all measured on a 6-point Likert scale (*Strongly agree, Moderately agree, Slightly agree, Slightly disagree, Moderately disagree, Strongly disagree*).

The next 3 questions consider the various types of medical data that can be stored by the NHS. Currently, the NHS keeps medical records which will include keeping track of any appointments you may have had with your GP. Please indicate how much you agree with the following statements.

- *I would feel uncomfortable with the possibility for the NHS to have my genetic data on record*
- *I feel uncomfortable with the possibility that the NHS have my sexual health data on record*
- *I feel uncomfortable with the possibility that the NHS have my mental health data on record*

2.3 Distribution & recruitment

We recruited participants by emailing a total of 390 community and volunteering groups across the UK, as well as science organisations, charities, and personal and academic contacts. A breakdown of these is in Appendix 3.4. Of those, 23 replied confirming their distribution of the survey, 11 declined, and 356 did not respond (response rate =8.7%). Posters were also distributed around libraries in Bristol, including Redland Library, Arts and Social Sciences Library, and Queens Library. Posters contained a QR code, so that the lead researcher could track the number of participants who completed the survey through this recruitment method. However, no participants were recruited via this method.

2.3.1 Obtaining a representative sample: Re-distribution

We aimed to recruit 300 participants. When I neared the halfway mark (*N*=145), I analysed participant demographics to assess the diversity of my sample. By comparing my data to UK census data (Office for National Statistics, 2011), I recognised that my most lacking demographics were: Asian participants (UK Census =7.5% vs. My data =2.7%); black participants (3.3% vs. 0.7%); participants with no formal qualifications (27% vs. 0%); and participants with GCSEs only (29% vs. 2.06%).

Therefore, as an attempt to obtain a more representative sample, I took a more targeted approach to recruitment. I got in contact with multiple BME (black and minority ethnic) student and campaigning groups (see recruitment table in Appendix 3.4). Of those, two responded and shared the survey with their group: SU BME Network and Union UCL BME Students' Network. However, no additional participants were obtained.

In addition, I used the survey recruitment site Prolific Academic (Prolific, 2014) and applied filters of no formal qualifications or GCSEs only. Through this method, I was able to obtain additional participants (*n*=53) who fulfilled these criteria.

2.4 Data analysis

Before data analysis commenced, data from Prolific Academic was combined with the original data to form one data set (*N*=430). An alpha level of .05 was used for all statistical tests.

2.4.1 Data cleaning

First, data were cleaned to eliminate those who did not finish the survey and/or give final consent (*n*=136, leaving *N*=294). Second, data from participants who finished the survey in under ten minutes (*n*=15) were eliminated as it was considered unlikely that they would have been able to read through all of the necessary information in that time, as the survey was estimated to take approximately 15–20 minutes. Finally, participants who were under 18 (*n*=3) and not from the UK (*n*=2) were excluded as they did not fit the research criteria (*N*=270).

2.4.2 Participant demographics

A total of 270 participants completed the study (170 female, 63%, Mean age =44 years). Approximately one-third of participants had at least a bachelor's degree (or equivalent), 4.8% of participants had a doctorate degree (or equivalent), and 17.4% of participants had GCSEs only (or equivalent). A large majority of participants were white (92.6%), and the remaining participants were Asian, biracial, or other ethnic group - 1.9% (UK Census data =7.5%), 0.7% (2.2%), and 4.1% (1%) respectively. There were no black participants (UK Census data =3.3%).

There was an almost equal divide between participants who had children (49.3%) and those who did not (50%), with the remaining data unavailable, i.e. participants selected 'Prefer not to say' (0.7%). Most participants had heard of genetic testing (93.3%) but did not work for the NHS (93%) or have genetics in their workplace environment (75.2%). Regarding their experience with genetic testing, 17.4% of participants indicated they, or someone close to them, had had a genetic test carried out by a doctor, but only 2.2% of participants had used an online genetic test to assess their genetic health risk. This statistic was slightly higher for use of online genetic tests to assess ancestry (6.7%). See Table 1 for further detail of participant demographics.

Table 1. Participant Characteristics in Quantitative Study.

Participant characteristics	Percentage of participants
Gender	
Male	35.9
Female	63
Data unavailable	1.1
Age	
18 - 25	23.4
26 - 35	11.1
36 - 45	8.5
46 - 55	23.5
56 - 65	14.6
66 - 75	8.5
76 - 85	4.8
Data unavailable	5.6
Highest level of school completed	
No schooling completed	1.5
GCSEs	17.4
A Level or equivalent	9.6
Trade/technical/vocational training or equivalent	3.7
Foundation degree or equivalent	1.5
Bachelor's degree or equivalent	32.6
Master's degree or equivalent	16.7
Professional training/Grad scheme or equivalent	12.2
Doctorate or equivalent	4.8
Ethnicity	
White/White British	92.6
Asian/Asian British	1.9
Biracial/Biracial British	0.7
Black/African/Caribbean/Black British	0
Other ethnic group	4.1
Data unavailable	0.7

Parent?	
Yes	49.3
No	50
Data unavailable	0.7
Works for the NHS?	
Yes	7
No	93
Last taught about genetics	
Still studying genetics	9.3
1 – 5 years ago	11.9
5 – 10 years ago	10
10 – 20 years ago	7
20+ years ago	21.1
Never taught	40.7
Genetics in workplace environment?	
Yes	16.7
No	75.2
Not in paid employment	7.8
Data unavailable	.4
Heard of genetic testing	
Yes	93.3
No	6.3
Data unavailable	.4
Had a genetic test by doctor (themselves or anyone close to them)	
Yes	17.4
No	81.9
Data unavailable	.7
Used online genetic test to assess genetic health risk	
Yes	2.2
No	97.4
Data unavailable	.4

Used online genetic test to assess ancestry	
Yes	6.7
No	93.3

Note. Data unavailable = Participants selected 'Prefer not to say'.

2.4.3 Scoring of measures

All scores were calculated using the mean score across items within each measure (biological knowledge, clinical knowledge, perception of knowledge, and acceptance of genomic medicine). I required participants to have completed 80% of the items in the measure in order to generate a score. As a final step, in order to make measures more easily comparable and equally weighted in the composites, all were rescaled so that the top score was equal to 1. Appendix 3.5 indicates the questions (*N*=13) that were eliminated from each section following reliability tests of Cronbach's alpha.

2.4.3.1 Biological knowledge of genetics

Participants' answers were scored (1 =correct, 0 =incorrect). For the questions 19–22, where participants were asked to give heritability estimates for various traits, all responses within 10% of the correct answer were scored as correct. This measure originally consisted of 25 items. However, question 5 was excluded because, as it was phrased, I realised there was more than one correct answer. Also, the measure did not reach acceptable reliability (α =.66). Inter-item correlation analyses revealed that 5 items were not worthy of retention and so were eliminated from the final measure (19 items, α =.74) (see Appendix 3.5).

2.4.3.2 Perception of genetic knowledge

Participants' answers to these 4 questions were scored to reflect their perceived level of knowledge (α =.87). For example, responses of '*Very good*' to the question '*When you hear the term DNA, how would you rate your understanding of what the term means?*' was given the highest score of 4. The final question in this measure was on a 5-point scale and so was rescaled so that the top score was equal to 4.

2.4.3.3 Clinical knowledge of genetics in healthcare

Again, participants' answers were scored (1 =correct, 0 =incorrect). For question 13, where participants were asked to indicate how much they had '*read or heard about genetic tests that predict the likelihood that a person will develop certain genetically influenced diseases or conditions'* on a 5-point scale from *'Not at all'* to *'Quite a lot',* answers were rescaled to be between 0 and 1. The original version of this measure had 17 items, however was deemed not reliable (α =.55). Removal of 8 items resulted in an increase in alpha (α = .64) (see Appendix 3.5).

2.4.3.4 Acceptance of genomic medicine

Participants' answers on the Likert scale questions were scored so that the top score of 5 was given to those who put '*Strongly agree'* with statements that implied a level of acceptance towards genomic medicine. For example, *'The benefits that the use of genetics in healthcare will bring to our healthcare system far outweigh any potential downsides*'. This scoring was reversed if participants put '*Strongly agree'* to statements that indicated a lack of acceptance, for example, *'The use of genetic data to inform medical decisions is overly intrusive'.* For question 1, which was on a 3-point scale, scores were rescaled so that the top score of 3 was equal to 5. The final two questions in this measure, which had three answer options, were scored as 1 and 0. A score of 1 was given to the answer that indicated higher acceptance of genomic medicine and a score of 0 was given to the remaining answer options (α =.83).

2.4.3.5 Additional measures

Three of the additional measures required scoring. Participants' level of comfort with the idea of their genetic data being held by the NHS was scored on a 5-point scale, with '*Strongly disagree'* on the statement '*I would feel uncomfortable with the possibility for the NHS to have my genetic data on record*' given the highest score of 5. This was repeated for level of comfort with both mental health data and sexual health data. The remaining additional questions were used as descriptive data.

2.4.4 Data screening

No variables had more than 10% missing data, so all were included in analyses. Scores were screened for outliers. Scores lower than .2 for acceptance (*n*=3) and biological knowledge

(*n*=1) were eliminated from further analyses. No other outliers were found with the remaining measures. Data was then tested for violation of assumptions of multiple regression. It passed tests of normality, linearity, and homoscedasticity (see Appendix 3.6 for results). To investigate multicollinearity between predictor variables, a Pearson correlation analysis was conducted. As can be seen in Table 2.1, the correlation between biological knowledge (*M* = .69, *SD* = .18) and clinical knowledge (*M* = .63, *SD* = .18) was .80, and thus violates this assumption. Therefore, these two predictor variables were combined to form a new variable – '*Combined knowledge'* (α =.89, *M* = .66, *SD* = .01).

Table 2.1 *Means, Standard Deviations, Minimum and Maximum Values, and Pearson Correlation Analyses Among Main Variables*

Variable	1	2	3	4
1. Biological knowledge	-			
2. Clinical knowledge	.80**	-		
3. Perception of knowledge	.55**	.51**	-	
4. Acceptance of genomic medicine	.06	.03	.15*	-
M	.69	.63	.56	.69
SD	.18	.18	.21	.17
Min	.21	.16	.11	.22
Max	1	1	1	1

Note. M = Mean. *SD* = Standard Deviation. Min = Minimum. Max = Maximum.

* $p < .05$. ** $p < .01$.

2.4.5 Statistical analysis: Multiple regression

2.4.5.1 The role of perception of knowledge

A second Pearson correlation analysis with all the final measures (i.e. including '*Combined knowledge'*) and all other variables revealed that higher scores on perception of knowledge were associated with higher scores on acceptance of genomic medicine (see Table 2.2). Acceptance was not correlated with combined knowledge. Perception of knowledge and combined knowledge were also positively correlated, $r = .55$, $p < .001$. However, this

correlation was not high enough to conclude that these two measures were conflated with one another.

The correlation results also provide support for the validity of my measures. As expected, both combined knowledge and perception of knowledge were positively correlated with education. In addition, both were positively correlated with being in a workplace that comes across genetic data and negatively correlated with the length of time since participants were taught about genetics. Also, combined knowledge was positively correlated with having heard of genetic testing prior to the survey and with previous use of an online genetic health risk test. Finally, perception of genetic knowledge was correlated with having had a genetic test carried out by a doctor.

The correlation results also give us some insight into my sample. The negative correlation between being female and being white indicates there were more women that identified as Asian, biracial, or other ethnic group than men. The negative correlation between age and working for the NHS shows that those participants who worked for the NHS were of a younger age than those who did not work for the NHS.

Table 2.2 Pearson correlation Among All Variables.

Variable	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
1. Combined knowledge	-														
2. Perception of knowledge	.55**	-													
3. Acceptance of genomics	.01	.15*	-												
4. Age	-.17**	-.16*	.03	-											
5. Being female	-.07	-.11	.00	.08	-										
6. Being white	.00	-.01	.06	-.05	-.12*	-									
7. Being a parent	-.25**	-.21**	-.01	.63**	.06	-.08	-								
8. Education	.41**	.28**	-.02	.14*	.08	.12*	.01	-							
9. Works for NHS	.07	.10	.06	-.14*	.12	.02	-.10	.06	-						
10.Time since taught genetics	-.39**	-.48**	-.05	.67**	.00	-.05	.51**	-.10	-.21**	-					
11. Genetics in workplace	.26**	.23**	.01	-.05	.10	.13*	.08	.46**	.23**	.23**	-				
12. Heard of genetic testing	.17**	.12	.01	.02	.02	.05	.04	.04	.02	.05	.06	-			
13. Genetic test by doctor	.10	.20**	.07	-.08	.06	-.01	-.03	-.03	.14*	.20*	.08	.11	-		
14. Online genetic health risk test	.13*	.04	.08	-.07	-.06	.07	.12	.12	.06	.03	.03	.03	.07	-	
15. Online genetic ancestry test	.06	.07	.07	-.03	-.05	-.01	.06	.06	.04	.03	-.05	.07	.11	.43**	-

Note. *p <.05. ** *p* <.01.

To explore further, a multiple regression was conducted to investigate the role of perception of knowledge (see Table 2.3). Model 1 indicates that perception of knowledge explained 2.4% of the variance in acceptance, $R2 = .024$. Perception of knowledge significantly predicted acceptance of genomic medicine (β =.16, p =.014).

In model 2, I adjusted for age and sex as control variables. Perception of knowledge remained a significant predictor variable (β =.16, p =.011). Neither sex nor age significantly predicted acceptance. Model 3 allowed us to control for all other covariates such as education, whether participants had heard of genetic testing prior to the survey and the time since participants were last taught genetics. Again, perception of knowledge remained a significant predictor (β =.17, p =.031). Finally, in model 4 I added combined knowledge as a control variable. Perception of knowledge still significantly predicted acceptance of genomic medicine (β =.21, p =.017). Combined knowledge did not predict acceptance (β =.023 p =.720) in either the unadjusted or adjusted models. A post-hoc power calculation revealed that I had 90% power to detect an effect size of 0.05 and 81% power to detect an effect size of 0.04 at a 95% confidence level, suggesting that if there was an actual effect of knowledge on acceptance, then the effect is small.

Table 2.3 *Enter Multiple Regression Results (Unstandardised B Weights, Standard Error, R_2 values, and P Values) for Acceptance of Genomic Medicine*

	β (95% CI)	*SE B*	*R_2*	*p*		*β (95% CI)*	*SE B*	*R_2*	*p*
Model 1					**Model 1**				
Perception of knowledge	0.16 (0.03, 0.23)	0.05	.024	.014*	Combined knowledge	0.02 (-0.10, 0.15)	0.07	.001	.720
Model 2 (adjusted for age & sex)					**Model 2 (adjusted for age & sex)**				
Perception of knowledge	0.16 (0.03, 0.24)	0.05	.027	.011*	Combined knowledge	0.03 (-0.10, 0.16)	0.07	.002	.673
Model 3 (adjusted for all demographics*)**					**Model 3 (adjusted for all demographics***)**				
Perception of knowledge	0.17 (0.01, 0.27)	0.07	.060	.031*	Combined knowledge	-0.01 (-0.16, 0.15)	0.08	.041	.918
Model 4 (adjusted for all demographics* & combined knowledge)**					**Model 4 (adjusted for all demographics*** & perception of knowledge)**				
Perception of knowledge	0.21 (0.03, 0.31)	0.07	.065	.017*	Combined knowledge	-0.09 (-0.26, 0.08)	0.09	.065	.293

*Note. * p < .05. ** p < .01.*

****Adjusted for: Age; Sex; Ethnicity; Parental status; Education; Works for the NHS; Time since last taught genetics; Genetics in workplace; Heard of genetic testing prior to survey; Genetic test carried out by a doctor; Used online genetic health risk test; Used online genetic ancestry test.***

2.4.6 Additional analyses

In addition to my main research question, I asked participants some additional questions to explore other interesting issues raised by genomic medicine (see section 2.2.1.7).

2.4.6.1 Consent procedure with genetically related family members

Figure 7.1 indicates the spread of responses given to the statement '*Family members share many genetic traits and may have the same genetic abnormalities associated with disease. Therefore, all immediate family members must give consent before an individual in that family*

gets a genetic test for medical reasons'. There is a clear pattern of responses, with most participants strongly (45.2%) or moderately (23.7%) disagreeing. Only 2.2% of participants strongly agreed. Overall, the majority of participants (83.3%) somewhat disagreed, with the remainder indicating that they agreed somewhat (16.6%).

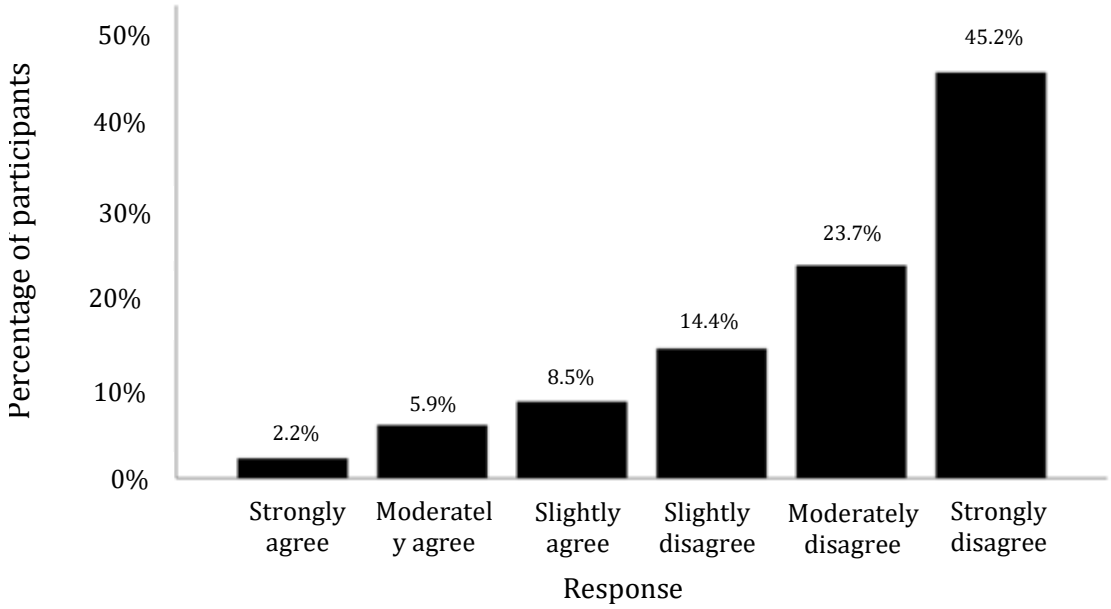


Figure 7.1 Bar chart indicating the spread of answers in response to the statement 'Family members share many genetic traits and may have the same genetic abnormalities associated with disease. Therefore, all immediate family members must give consent before an individual in that family gets a genetic test for medical reasons'

2.4.6.2 Is my destiny in my genes?

Figure 7.2 shows the spread of responses to the statement '*I believe that my destiny is written in my genes'.* The most popular response was 'Slightly agree' (28.3%), followed by 'Moderately disagree' (21.9%) and 'Strongly disagree' (19%). There was an almost even split between somewhat agreeing (47.6%) and somewhat disagreeing (52.4%) with the statement.

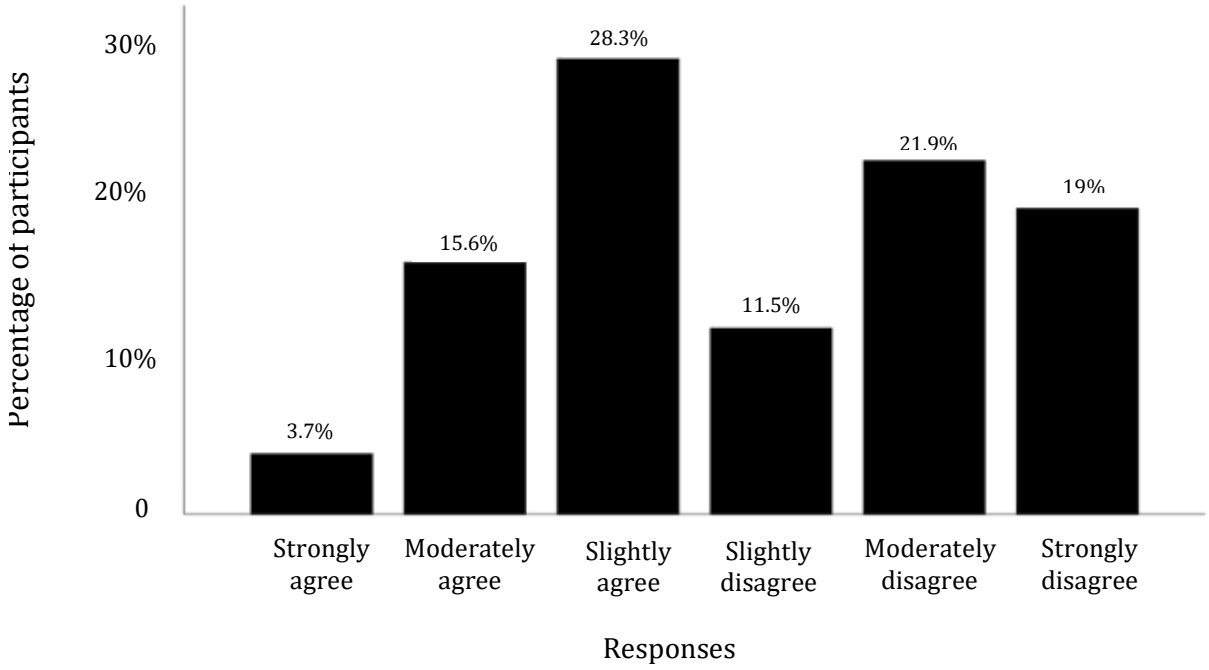


Figure 7.2 Bar chart indicating the spread of answers in response to the statement 'I believe that my destiny is written in my genes'.

2.4.6.3 Is prevention better than cure?

Figure 7.3 shows participants' responses to the statement '*Preventing health problems is preferable to curing health problems*'. The majority of participants (73.4%) strongly agreed with this statement, 18.9% moderately agreed and 5.9% slightly agreed. Overall, a large majority (98.2%) of participants somewhat agreed with this statement, with the remaining 1.8% stating they slightly disagreed (1.1%) or strongly disagreed (0.7%).

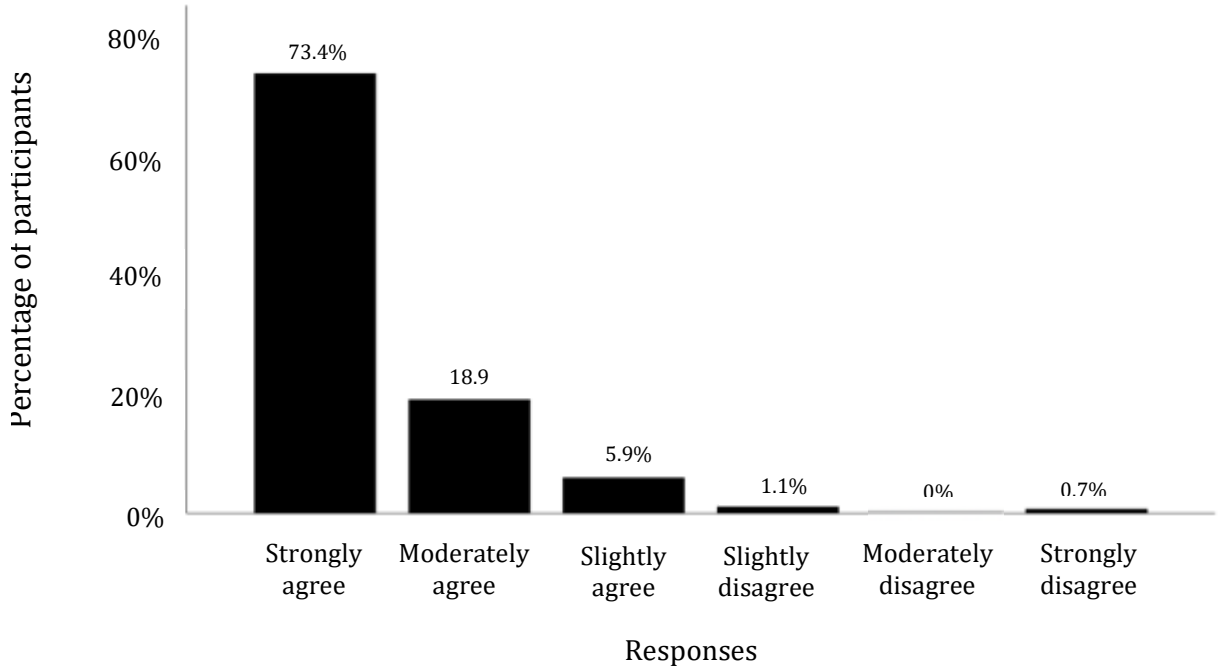


Figure 7.3 Bar chart indicating the spread of answers in response to the statement 'Preventing health problems is preferable to curing health problems'

2.4.6.4 Genetic exceptionalism: Do people have less trust in the NHS storing their genetic data than their mental or sexual health data?

Pearson correlation analyses revealed correlations between feelings of comfort with the three different types of data (genetic, mental health, and sexual health) being held by the NHS (Table 2.6). The highest correlation was between mental health data and genetic data, followed by mental health data and sexual health data, and then genetic data and sexual health data.

Table 3 *Means, Standard Deviations, Minimum and Maximum values, and Pearson Correlations Among Comfort with Different Types of Data Being Held by the NHS*

Variable	**1**	**2**	**3**
1. Comfort with genetic data in NHS	-		
2. Comfort with sexual health data in NHS	.61**	-	
3. Comfort with mental health data in NHS	.74**	.67**	-
M	.72	.71	.73
SD	.28	.28	.28
Min	0	0	0
Max	1	1	1

Note. M = Mean. *SD* = Standard Deviation. Min = Minimum. Max = Maximum. * $p < .05$. ** $p < .01$.

Figure 7.4 shows the spread of responses for each type of data. For each data type, there was a similar distribution of responses. For example, 3.7% of participants responded with 'Strongly agree' for genetic and sexual health data, with a similar 2.2% for mental health data. Overall, 79.2% of participants somewhat disagreed (i.e. a combination of all disagree responses) with the statement *'I would feel uncomfortable with the possibility for the NHS to have my genetic data on record',* indicating that these participants felt comfortable with the idea of the NHS having their genetic data. For mental health data, the proportion of participants that felt this way was slightly less (77.4%), and slightly less again for sexual health data (75.6%).

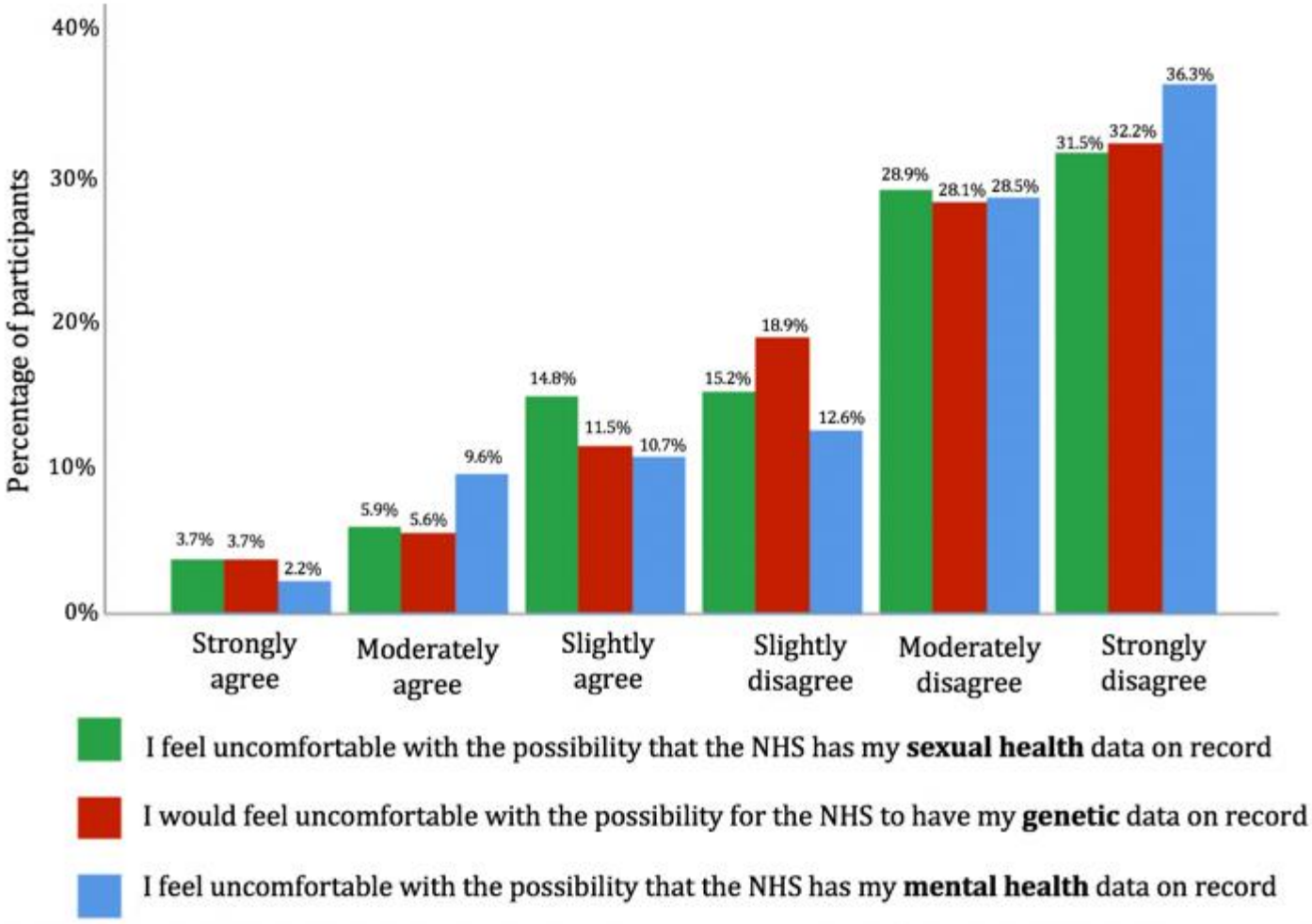


Figure 7.4. Bar chart indicating participants' responses as to how comfortable they would feel with different types of data (sexual health, genetic, & mental health) being held by the NHS.

2.4.6.5 Specific questions with lowest and highest scores

In order to explore the specifics of participants' knowledge, the knowledge questions (in the combined knowledge measure) with the lowest scores (more than 60% of participants with incorrect answer) and the highest scores (more than 85% of participants with correct answer) were calculated. For the acceptance measure, the two questions that instigated the least and most accepting responses were calculated.

2.4.6.5.1 Knowledge questions with lowest scores

Table 4.1 below indicates the questions that participants had the most difficulty with. In particular, participants struggled to define the term 'heritability'. In line with this, the majority

of participants did not get within 10% of the correct answer when estimating the heritability of both weight and school achievement. Finally, 60.7% participants did not know that there are approximately 20,000 genes in the human genome.

Table 4.1 *Table of the Questions with Highest Amount of Incorrect Responses, and the Percentage of Participants with the Incorrect Answer.*

Question	Percentage of participants with incorrect answer
The word 'heritability' means… • The proportion of the variation in the physical composition of a population accounted for by genetic variation • The proportion of a physical composition that is passed on to the next generation • The proportion of a person's characteristic that is accounted for by genes • The proportion of genes that are important for the development of a characteristic	83.3%
Approximately how many genes does the human DNA code contain? • 2,000 • 1 million • 3 billion • 20,000	60.7%
On a scale of 0-100 how important do you think genetic differences are between people in explaining individual differences in the following traits (with 100 = only genetic differences can explain individual differences in traits): WEIGHT	78.5%
On a scale of 0-100 how important do you think genetic differences are between people in explaining individual differences in the following traits (with 100 = only genetic differences can explain individual differences in traits): SCHOOL ACHIEVEMENT	78.1%

Note. Green = the correct response. Red = the most common incorrect response.

In addition, only 28.9% of participants had heard of the 100,000 Genomes Project, and even fewer (14.4%) had heard of Genomics England.

2.4.6.5.2 Knowledge questions with highest scores

Table 4.2 shows the questions that most participants answered correctly. For example. nearly all participants understood the mechanism of a one in four chance of a child having a genetic disease, (95.9% of participants) and a large majority of participants (93.7%) were aware that the study of genetics can lead to better treatments.

Table 4.2 *Table of the Questions with Highest Number of Correct Responses, and the Percentage of Participants with the Correct Answer.*

Question	Percentage of participants with correct score
A doctor tells a couple that they've got a one in four chance of having a child with genetic disease, This means... • ...if their first three children are healthy, the fourth will have the illness • ...if their first child has the illness, the next three will not • ...each of the couple's children will have the same risk of suffering from the illness • ...if they have only three children, none will have the illness	95.9%
What is the main function of all genes? • Storing information for protein synthesis • To provide energy to the cell • To clear out waste from the cell • To repair damage to a cell	92.2%
A single gene can influence several different traits or diseases	85.9%

- True
- False

Which of the following is FALSE: • By eating a genetically modified fruit, a person's genes could also become modified • The cloning of living things produces genetically identical copies • All plants and animals have DNA	87.1%
There are many common diseases where the study of genetics can show the road to better treatment • True • False	93.7%
The use of patients' genetic data to deliver targeted therapies is already changing people's lives • True • False	92.2%
The rapidly expanding role of genetics in many healthcare decisions is already increasing the demand for qualified genetics professionals • True • False	94.1%
Currently, the NHS has every individual patient's genetic code stored on their database • True • False	96.7%

Note. Green = correct response.

2.4.6.5.3 Acceptance statements with lowest and highest score

The highest possible score on each acceptance statement was 1, and the lowest 0, with 4 intermediate scores in between. Thus, if every participant (N=270) responded with the most accepting answer on a statement (i.e. giving them a score of 1), the overall score for that

statement would equal 270. From this, I could calculate which statements instigated the most and least accepting responses in terms of participant attitude to genomic medicine. The statement that received the least amount of acceptance was '*I am concerned about who will have access to my genetic data once it is stored within an NHS database*' - agreeing with this statement indicated a lack of acceptance. This statement scored 124/270, and Figure 7.5 demonstrates the spread of responses. Importantly, the arrows on the figure show that this statement instigated fewer accepting responses than non-accepting responses.

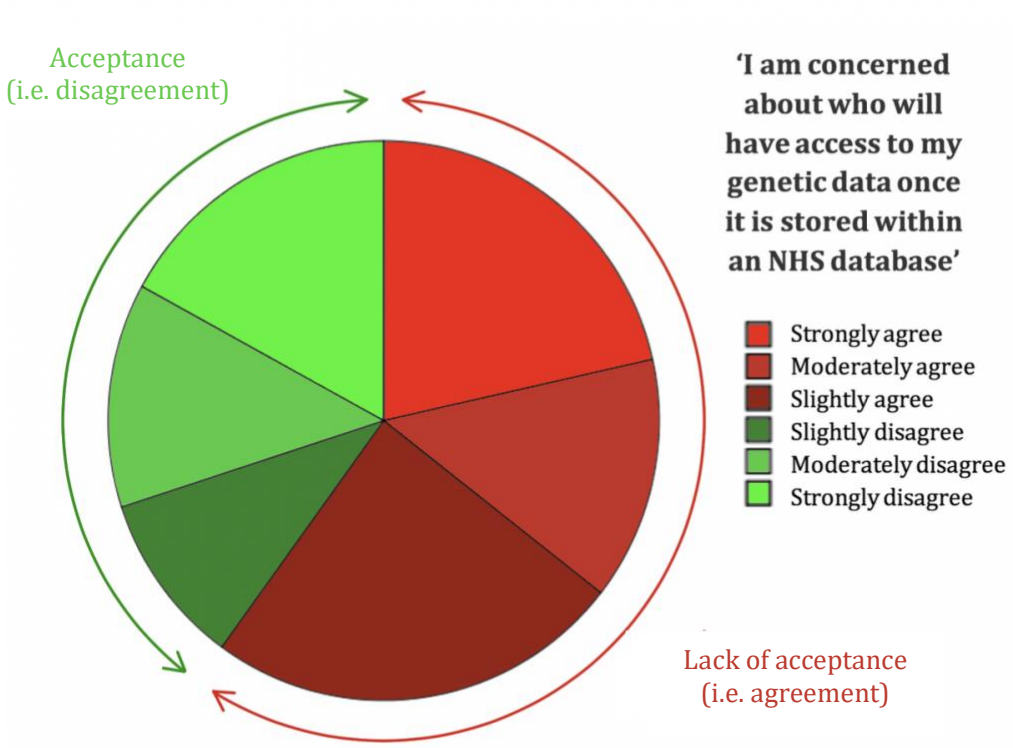


Figure 7.5 Pie chart indicating the spread of responses to the statement 'I am concerned about who will have access to my genetic data once it is stored within an NHS database'

Contrastingly, the statement with the highest score of acceptance was '*If I was told that knowledge of my genetic data may improve the effectiveness of a medical intervention that I required, I would not hesitate to have my genetic data tested'.* This statement scored 240/270 - agreeing with this statement indicated acceptance - and the spread of responses can be seen in Figure 7.6. Crucially, the arrows show the large difference between the amount of accepting responses versus non-accepting responses.

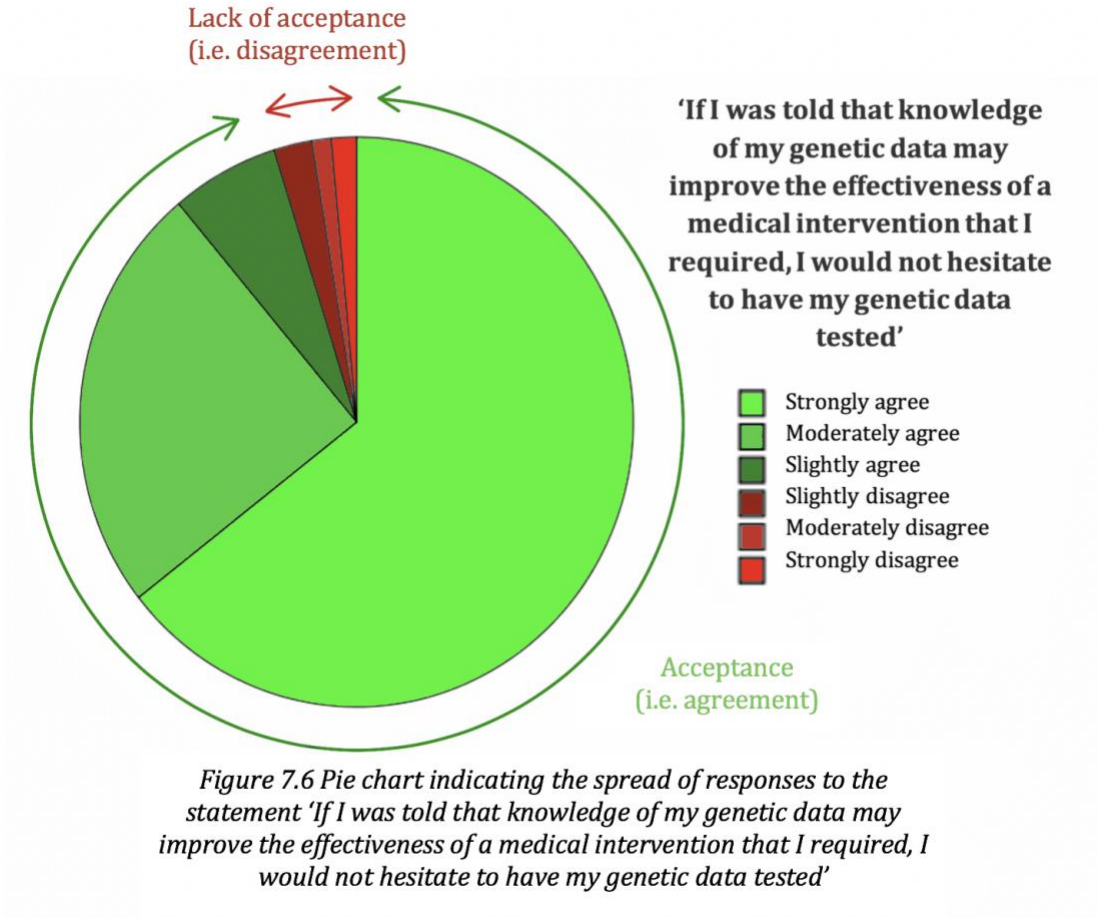


Figure 7.6 Pie chart indicating the spread of responses to the statement 'If I was told that knowledge of my genetic data may improve the effectiveness of a medical intervention that I required, I would not hesitate to have my genetic data tested'

2.5 Discussion

In this chapter I explored the predictive power of different types of genetic knowledge, i.e. biological, clinical, and perception of knowledge, on the acceptance of genomic medicine. My study found that UK participants' knowledge of genetics (a combination of biological and clinical knowledge, i.e. combined knowledge) did not predict their acceptance of genomic medicine. However, participants' *perception* of their genetic knowledge predicted their acceptance but with a small effect size, explaining only 2.4% of the variance. Thus, the findings in this study do not support the hypothesis that fear of genomic medicine is due to a lack of understanding of genetics. Nonetheless, the findings provide some evidence that members of the public who *perceive* themselves to be more knowledgeable about genetics are more accepting of genomic medicine.

It is also important to note that, on average, participants scored 66% in the combined knowledge measure. This is almost identical to the average score of 65.5% in Chapman et al's (2019) recent study, which the authors described as 'poor' given the multiple-choice format of questions which greatly increases peoples' chances of getting the correct answer. Given that my questions were also in multiple-choice format, and that a large majority of my sample were highly educated - 66.3% were educated to bachelor's degree level and above - I can come to a similar conclusion and suggest that genetic knowledge amongst my sample was poor. This is concerning because the mainstreaming of genomic medicine means genetics will become more relevant for people in the UK over the coming years; having an understanding of basic genetic concepts is likely to help individuals navigate their way through this complex area of medicine.

Whilst this study cannot ascertain cause and effect, I controlled for multiple variables such as age, sex, parental status, and experience with genetic testing. The role of perception of knowledge remained statistically significant. Further, whilst participants' combined knowledge was positively correlated with their perception of their genetic knowledge, the predictive power and significance of perceived knowledge also remained when combined knowledge was a control variable. Therefore, independent of how knowledgeable the participants were, their perception of their knowledge was more predictive than their actual knowledge. The following section will explore these main findings in more detail.

2.5.1 Present study: *perception* of knowledge predicts acceptance of genomic medicine

Previous studies have found factors such as experience with genetic disease (Henneman, Timmermans, & Wal, 2006) or trust in science and technology (Gottweis, 2002) are more important than genetic knowledge as a predictor of acceptance. Contrastingly, the present study indicates that participants' *perception* of their genetic knowledge was the most important predictor. In my study, participants who were more confident in their genetic knowledge were more accepting of genomic medicine. This result makes intuitive sense: feeling more confident in your genetic knowledge may also help you feel more prepared for any issues that may arise. This would be particularly comforting given that there are so many unknowns in genomics. This is in line with previous research that shows having more confidence in your own abilities is a powerful tool for long-term health behaviour change

(Strecher et al. 1986), which could include changing behaviour in response to genetic results. This is particularly important for a smooth transition to difficult changes to health practice - genomic medicine represents such a difficult change for many members of the public.

To interpret this result further, it is important to also consider the relationship between perceived and actual genetic knowledge (i.e. combined knowledge). Participants displayed a level of accuracy when estimating their level of knowledge, however there was room for improvement. This is consistent with previous research that indicates people can be inaccurate when estimating their level of knowledge (West & Stanovich, 1997). Lanie et al. (2004) attributed these inaccurate estimations to increases in exposure to genetic terms from the media. Indeed, there have been calls for scientists to advocate more for correct reporting of results and the encouragement of realistic understandings of science (Evans et al. 2011). For example, it has been suggested to replace the use of the phrase 'gene versus environment' with 'gene-environment interaction' (Condit, 2007). Encouragement of the media to use terminology that will foster correct understandings of genetics is particularly vital because the public get most of their information about genomics from the media (Parry, 2019).

2.5.2 Present study: no evidence for an association between genetic knowledge & acceptance of genomic medicine

Our finding of a null relationship between genetic knowledge and acceptance of genetics in healthcare is in line with findings from previous research (Henneman, Timmermans, & Wal, 2006; Sturgis, Brunton-Smith, & Fife-Schaw, 2010). This suggests that efforts to educate the public with scientific facts about genetics may not have the deficit model's predicted effect of increasing public acceptance of genomic medicine.

In turn, my findings go against the notion of a deficit model (Sturgis & Allen, 2004) and findings of a positive relationship (Allum et al. 2014; Chapman et al. 2019; Human Genetics Commission, 2001; McClintock, 2019; Pardo et al. 2002) as well as findings of a negative relationship (Jallinjoa & Aro, 2000). It may be that the deficit model is incorrect, and that people's knowledge of genetics is not associated with their acceptance of genomic medicine. Or, given how quickly the field of genetics in healthcare is changing and that many more people

may have had experience with genetic testing, the deficit model may be out-of-date. However, it is important to consider alternative reasons for my finding, as discussed below.

2.5.2.1 Use of formal scientific knowledge

Despite a belief amongst some scientists and policy-makers that 'to know it [science] is to like it' (Bauer, Petkova, & Boyadjieva, 2000, pg. 42), the present study failed to reveal such a finding. However, it is important to point out that 'loving' science and being sceptical about science and scientific research are not mutually exclusive. Indeed, one can enjoy and appreciate the scientific process but have an awareness of its disadvantages and failings. Regardless, my measure of genetic knowledge, both biological and clinical, was based on the testing of factual scientific knowledge. Other studies that found a positive association also tested factual scientific knowledge (Allum et al. 2014; Human Genetics Consortium, 2001; McClintock, 2019). This automatic framing of scientific knowledge in such a general, formal way may be overlooking other aspects of scientific knowledge that are equivalently, or indeed more, important. Actually, lay understandings of science can be detailed and sophisticated without possessing formal scientific knowledge (Sturgis, Cooper, & Fife-Schaw, 2005). For example, institutional knowledge, i.e. knowledge of the regulatory framework of science, which includes knowledge of the rules and methods involved in data storage and de-identification, as well as how data is shared within and across institutions, has been suggested as most important in establishing trust in science (Bauer, Petkova, & Boyadjieva, 2000). Given the importance of trust in public acceptance of scientific technology (Davies, 2017; Gottweis, 2002; Open Data Institute, 2018; Siegrist, 2000), it could be reasonably concluded that institutional knowledge is most important in improving public acceptance of genomics. Thus, overall, my conceptualisation of 'knowledge' as the understanding of the biological mechanisms of genetics and the current state of genetics in healthcare may have overlooked other aspects of the public's complex and nuanced understandings of genetics. Bucchi & Neresini (2008, pg. 60) summarised this point, '*Lay knowledge is not an impoverished or quantitatively inferior version of expert knowledge; it is qualitatively different.'* Future research could include measures of participants' institutional knowledge to explore its role in public acceptance of genomic medicine.

2.5.2.2 Controversial nature of genomics in UK

Alternatively, my results may be due to Bak's (2001) finding that the relationship between knowledge and acceptance is weaker with more controversial scientific technologies. The UK is the world leader in genomic medicine - we have progressed more than any other country in making the promise of personalised medicine a reality. Thus, it may be that the UK public perceive genomics to represent a threat in a way that other countries do not. In turn, the relationship between knowledge of genetics and acceptance of genomic medicine may be weakened so much that it ceases to exist.

2.5.2.3 Differences in measures across studies

Finally, the difference between my findings and the findings of studies that show a significant relationship between knowledge and acceptance may be a result of the different measures used. In my study, I used a combination of survey questions from multiple sources as well as newly constructed questions based on research into common misunderstandings of genetics (see 2.2.1 for details of survey development). The development of the measure of knowledge was based on both biological and clinical knowledge of genetics. To the best of my knowledge, no other study has looked at clinical knowledge of genetics in UK healthcare. The measure was validated through its positive associations with education, being in a workplace that comes across genetic data, and familiarity and experience with genetic testing, as well as its negative correlation with the length of time since participants were taught about genetics.

Studies with results that differed to mine used different measures of knowledge. For example, the Human Genetics Commission's (2001) assessment of participants' knowledge was less extensive; participants were judged only via their ability to appropriately classify 4 characteristics as completely inherited, e.g. cystic fibrosis. Also, McClintock's (2019) study was an experiment that measured the short-term effect of an intervention using PUGGS (Carver et al. 2017). In contrast, my study was non-experimental and whilst I incorporated some of the same questions from the PUGGS, my final measure was substantially different.

Overall, due to the diverse measures used, the different studies may have captured similar yet subtly different aspects of genetic knowledge. In turn, the relationship between knowledge and acceptance may have been affected. However, whilst plausible, this reasoning implies that the

investigated relationship may not have sufficient strength to withstand variation in how it is measured.

2.5.3 Additional findings

In addition to my main research questions, I explored important questions that are raised by the continued advancement of genomic medicine in the UK. Below is an exploration of my findings.

2.5.3.1 Genetic exceptionalism: Do people have less trust in the NHS storing their genetic data than their mental or sexual health data?

A large majority of participants (79.2%) indicated they felt comfortable with the NHS having their genetic data on record. This statistic was slightly higher than the proportion of participants who felt this way about mental health data (77.4%) and sexual health data (75.6%). Further, feelings of comfort with genetic data correlated highly with feelings of comfort with both sexual health data and mental health data. This particular finding, i.e. in the context of the NHS having a record of their genetic data, does not provide support for the concept 'genetic exceptionalism', i.e. the perception that genetic data is different to other types of medical data (Davies, 2017). Given that the UK is leading the way with genomic medicine, perhaps UK residents are becoming more familiar with genomics and consequently becoming more receptive to the idea of genomic medicine.

However, this finding may simply be reflecting the high levels of trust and confidence people have in the NHS and doctors (Ipsos MORI, 2013). Such automatic and large feelings of trust in an organisation like the NHS may influence individual judgement on certain issues, which could include storage of their genetic data. Indeed, future research should explore the public's perception of genetic data being shared with research institutions, commercial companies, or with healthcare organisations outside of the UK. Such future work could also explore whether the large-scale public trust in the NHS can have a *negative* impact on individual choice and judgement. For example, participants could be presented with a scenario in which there is a clear answer as to the most appropriate course of action. However, one group could be given this scenario in an NHS context and the other group in the context of a commercial company. Differences in responses could reveal whether trust in the NHS can cloud judgement.

2.5.3.2 General opinion: genetic relatedness between family members & the implications for consent

A large majority of participants (83.3%) disagreed with the statement 'all immediate family members must give consent before an individual in that family gets a genetic test for medical reasons'. This question is linked to one of the main difficulties within genomics – genetic relatedness between family members. Perhaps participants believed this would be impractical and too difficult to achieve. Alternatively, participants may have felt that personal choice and autonomy when making medical decisions is more important than obtaining permission from those it may affect. It could also indicate the importance that people attach to maintaining patient confidentiality – asking all family members for consent may reveal private information about an individual's medical status. Future research could use qualitative methods to explore this finding in more depth in order to provide useful recommendations for practice and patient-doctor relationships.

2.5.3.3 General opinion: genetic determinism

Interestingly, 47.6% of the participants indicated they believed their 'destiny is written in my [their] genes'. Therefore, nearly half of the participants displayed beliefs consistent with genetic determinism. This is concerning, particularly given that one-third of our sample has at least a bachelor's degree. This is concerning because it could affect how these individuals may interpret genetic results they are given. Given the reputation of the media in constructing beliefs of genetic determinism (section 1.6.2.4.1), this finding suggests we should increase our efforts to ensure that the media are correctly reporting genetic results and using language that does not foster the out-of-date idea that behavior is always caused by *either* nature *or* nurture.

2.5.3.4 General opinion: prevention versus cure

Nearly all participants (98.2%) indicated they believed that preventing health problems is preferable to curing health problems. Genomic medicine's key aim is to understand more about disease *before* it develops (Caulfield, 2019). Researchers are striving for the identification of individuals at risk of disease, so that they can administer necessary treatment or precautionary measures before the development of disease. Thus, in the eyes of genomic medicine, prevention is preferable to cure. So, this finding indicates nearly all participants support the

main ethos of genomic medicine, possibly without being consciously aware of it. Future research could explore whether increased communication of this message improves acceptance of genomics.

2.5.3.5 The specifics of knowledge

A large majority of participants demonstrated that they understood the basics of genetics, such as the fact that a single gene can influence several different traits or diseases. Where participants struggled, however, was with the definition and application of heritability estimates and, similar to Chapman et al's (2017) study that found less than 50% of participants were aware of the number of genes in DNA, 60.7% of the participants in my study also failed to answer this correctly. Knowledge of the public's weaker areas of genetic knowledge can help to guide education efforts.

2.5.3.6 The specifics of acceptance

Finally, my additional analyses revealed that the statement with the lowest score of acceptance was 'I am concerned about who will have access to my genetic data once it is stored within an NHS database'. This contradicts my aforementioned result that 80% of participants were happy with their data being stored on an NHS database, and confirms the need for further research. Contrastingly, the statement with the highest score of acceptance was 'If I was told that knowledge of my genetic data may improve the effectiveness of a medical intervention that I required, I would not hesitate to have my genetic data tested'. Overall, these results indicate that participants were most concerned about third party access to their data but would also have a genetic test if it meant improving a required treatment. Future research could explore whether participants feel their concerns outweigh their perceived benefits and future NHS consent materials should be very clear about whether or not the data will be used by third parties.

2.5.4 Limitations

Our study provides important and novel evidence that higher confidence in genetic knowledge is associated with increased acceptance of genomics, but it is not without limitations. First, I did not manage to recruit enough Asian, biracial, or black participants. This is important as people of colour are more concerned about the rise of genomics leading to racial

discrimination (Goldenberg et al. 2011; Zimmerman et al. 2006). Therefore, I am likely to have missed an important perspective that may have influenced results. Further, participants who were Asian or biracial were also more likely to identify as female. This means the data was lacking in Asian and biracial men, and that my sample is likely to have overrepresented the white male perspective. Further, those in my sample who worked for the NHS were also more likely to be younger. Again, this is important because I may have missed influential opinions of those who have more experience working in the NHS. However, this was not a survey aimed at individuals working in the NHS and the opinion of healthcare professionals specifically was beyond the scope of this project.

Second, my survey was framed so that participants were asked biological and clinical questions first and their perceptions of knowledge second. This may have resulted in ratings of perception that were not truly representative of participants' confidence in their knowledge in their everyday life as it may have knocked the confidence of some but boosted the confidence of others, particularly as the survey was designed to be difficult. However, I did not give participants feedback. So, arguably, this may have actually provided *more* accurate ratings of confidence as they would have been a reflection of participants' actual knowledge as well as their confidence. Those who felt they got more correct, i.e. due to both higher confidence in their knowledge as well as better actual knowledge, were likely to consequently give higher ratings of confidence. Further, my measure of confidence in knowledge may actually reflect participants' conscientiousness or perfectionism.

Third, it is important to consider that my measure of perception of genetic knowledge may reflect something other than what it intended to measure. For example, estimates may actually indicate participants' *broader* level of confidence and/or self-esteem; more confident participants with higher self-esteem, regardless of their level of confidence specific to their genetic knowledge, may have given themselves higher scores on the perception of knowledge measure than their less confident counterparts. Indeed, other studies that explore genetic knowledge include measures of personality as it is established that this can affect your attitudes towards genomic medicine (Chapman et al. 2019). Therefore, future research should include a personality measure in order to control for the effects of general confidence on confidence in genetic knowledge. We could then establish whether acceptance is linked to subject-specific confidence or confidence more generally.

Finally, previous research indicates the importance of social class in shaping people's opinion on genomics (Sturgis, Cooper, & Fife-Schaw, 2005). However, I did not include a measure of social class, and thus were not able to explore its role in shaping either people's opinion or their knowledge. However, I did attempt to obtain a representative spread of educational levels and this in part makes up the social class scale.

2.6 Chapter summary

In this chapter, I designed, distributed, and collected data using a survey that explored participants' biological knowledge of genetics, clinical knowledge of genetics in healthcare, perception of their genetic knowledge, and their acceptance of genomic medicine. My results indicated that neither biological nor clinical knowledge predicted participants' acceptance of genomics. Participants' perception of their genetic knowledge was found to predict acceptance, but with a small effect size. Whilst it may be that the deficit model is incorrect or out-of-date, I also considered alternative reasons for my finding. This chapter also considered some additional findings and the implications for future research.

Chapter 3: Qualitative study – An exploration of an informed public's opinion of genetics in healthcare

3.1 Introduction

Genomic medicine is a controversial topic. Public concerns include: the use of genetic data by insurance companies (Cook, 1999; Geer et al. 2001; Hall et al. 2005; Lemke et al. 2010; Stockdale, Cassell, & Ford, 2018), employers (Geller et al. 2002; Tambor et al. 2002), or commercial companies (Hapgood et al. 2004; Middleton et al. 2019; Trinidad et al. 2010), the use of genetic data for reasons other than their own medical benefit, e.g. discrimination; the potential for damages to medical privacy from sharing their genetic data for research (Anderlik & Rothstein, 2001; Gill & Richards, 1998; Kaufman et al. 2009); and feeling that their data is not safe and secure within the NHS (Stockdale, Cassell, & Ford, 2018). These concerns are discussed in more detail in section 1.7.

Ultimately, these anxieties stem from the unique nature of genetic data which challenges conventional practices of consent and confidentiality (see section 1.5.3 for more detail). Firstly, regarding confidentiality, genetic test results for one individual have the potential to affect a multitude of others who share DNA. Confidentiality is tested further by the fact that a patient must share their phenotypic data (i.e. their medical records) to ensure their genetic data is clinically useful. Secondly, the prospect of discovering genetic variants of diagnostic significance not linked to the primary reason for the genetic test raises difficulties with consent (Mackley & Capps, 2017). Overall, these distinct features of genetic data arguably make it stand out from other types of health data. Indeed, this may help to explain the reasons behind 'genetic exceptionalism', whereby people feel genetic data is categorically different to other types of data (Davies, 2017). In Chapter 2, my quantitative study did not find any evidence for this (see section 2.5.3.1), however may have been confounded by high levels of trust in the NHS (REF). Further qualitative exploration is needed to provide more insight into genetic

exceptionalism amongst the UK public and to unpick exactly what it is about genetic data that may foster feelings of unease amongst the general public.

These complex concerns have not gone unnoticed by the scientific or medical community; an open dialogue has been established to provide a space for the discussion of ethical issues. For example, the former 'European Meeting on Psychosocial Aspects of Genetics' (EMPAG) has, since 2019, been added to the larger 'European Society of Human Genetics' conference, indicating the recognition that it needed to become part of the broader conversation about genomics (European Society of Human Genetics, 2018). Further, in the Chief Medical Officer's 2016 annual report entitled 'Generation Genome', an entire chapter was dedicated to the ethics and the implications for the NHS (Davies, 2017), and the Wellcome Genome Campus has a whole department dedicated to society and ethics research (Wellcome Genome Campus, 2019). In fact, recognition of these issues dates back to the Human Genome Project, where 3-5% of the annual budget was set aside for the exploration of ethical, legal and social issues (Sansgiry & Kulkarni, 2003).

However, whilst these initiatives are admirable, more research is required to deepen our understanding of the public's perception both of the ethical issues in genomics and of genomic medicine more broadly. This will help to inform our responses to the difficulties that arise from the use of genetic data in healthcare. Given the aforementioned complex and controversial nature of genomic medicine, qualitative research may offer a more useful contribution to this work; it allows a depth of insight and may help to tease out complexities in a way that is not possible in quantitative research, as well as possibly providing public led suggestions for future healthcare practices.

Thus, the present study sought to deepen our understanding of the UK public's perception of genetics in healthcare (the importance of research into the UK public is discussed in section 1.9.2). I conducted two focus groups to explore public opinion and thoughts on the use of genetic data in healthcare. Unlike previous qualitative research (Bates et al. 2005; Stockdale, Cassell & Ford, 2018; Waters, Ball, & Gehlert, 2017), participants in my study were given information about the current state of genomic medicine in the UK. This was in order to gather opinion about genomic medicine as it exists now, as opposed to opinions based on false information and/or ideas. Overall, this was an exploratory study with no specific hypothesis,

but an aim to provide findings that may help to guide communication and public engagement efforts within genomic medicine.

3.2 Methods

We used focus groups to obtain a broad range of detailed information which I analysed using the General Inductive Approach (Thomas, 2003).

3.2.1 Ethical approval & informed consent

Ethical approval was granted on 9th April 2019 from the University of Bristol School of Psychological Sciences Research Ethics Committee (SREC) (see Appendix 4 for details).

Participants received Participant Information Sheets (PIS) in advance of the focus group. This informed the participants that the conversation would be recorded and transcribed by a transcription service, and that the data would be kept anonymous as names would not be transcribed. Paper consent forms were given to participants upon arrival, that assured participants of the confidential nature of the data. Participants were given as much time as they required in which to decide to consent. Before commencing the focus group, the lead researcher ensured all participants had read the PIS and the consent form, and that they were given an opportunity to ask questions. The lead researcher then gave participants a verbal overview of how the session would go forward. Written consent was obtained from all participants.

3.2.2 Data protection & confidentiality

The completed consent forms and participant demographic information sheets were stored in separate, safe and secure locations by the lead researcher. Recordings were made on university-owned dictaphones. The audio recordings were sent to University Transcriptions (www.universitytranscriptions.co.uk) and were all checked against the recordings by the lead researcher to check that technical language had been transcribed correctly. After transcription, the audio recordings were deleted. All identifiable information (i.e. names) were removed and replaced with participant ID numbers. The anonymised transcripts were stored separately from the consent forms and participant demographic information sheets so that no link could be made.

3.2.3 Participant identification & recruitment

Convenience sampling was used. Participants had to be over 18, a UK resident, and have English as their first language or an equivalent level of fluency. Posters displaying the details of the study, the reimbursement amount (£10), and the contact information of the lead researcher were distributed around Bristol. Distribution locations included:

- Cotham Pharmacy & Post Office, Cotham
- Redland Library, Redland
- Arts and Social Sciences Library, University of Bristol
- The Canteen, Stokes Croft
- Café Kino, Stokes Croft
- The Arts House, Stokes Croft
- Salvation Army, Stokes Croft
- Tuck News, Cotham
- The Cotham Arms
- St Peter's Hospice, Stokes Croft

Potential participants expressed interest via email. The lead researcher responded by sending information about the structure of the focus group and a copy of the PIS. Participants were also asked for basic demographic information (age, sex, & ethnicity). Recruitment continued until 16 participants who met the inclusion criteria (2 x focus groups of 8 participants) were confirmed. A week before each focus group, each enrolled participant was sent a reminder. All but one of the participants attended the focus groups (N = 15).

3.2.4 Data collection & focus group conduct

The focus groups were conducted in the School of Psychological Science common room. The lead researcher led the focus group, and Dr Robyn Wootton was also present to make notes about body language and other details not captured by the recording. Participants were first given a brief presentation about the current state of genetics in healthcare, using information

from the NHS (2016). This began with the showing of an approximately 6-minute clip 'Whole Genome Sequencing and You' (Icahn School of Medicine, 2012). This video detailed: the biological structure and function of DNA and proteins; the technology of whole genome sequencing; that genetic variants and the environment can influence your risk of developing common diseases; that variants in your genome can influence your response to medication; that specific genetic variants can lead to the development of serious and/or rare disease; and the existence of variants of unknown significance. This clip was chosen as, in the judgement of the lead researcher, it gave a balanced view of genetics in healthcare and accurate information. This clip was followed by a 5-minute presentation from the lead researcher, that detailed: the timeline of genomic medicine; what the NHS currently tests for, i.e. monogenic disorders, cancer, and high penetrance mutations; and how developments in research may lead to a system of 'personalised medicine' in the NHS. I did not discuss issues such as data protection and privacy in this introduction. Conversation was then prompted by questions from the lead researcher, which were both displayed on the presentation slides and read out verbally. A list of these questions can be seen in Appendix 5. The presentation slides can be seen in Appendix 6.

The questions used were largely adapted from the 'Acceptance of genetics in healthcare' section of the survey (see section 2.2.1.6). Additional questions such as: '*Would you have your genetic data tested by the NHS if you were trying for a baby and wanted to know your carrier status?*' and '*Would you have your genetic data tested by the NHS if you were at risk of carrying a high penetrance mutation?*' and '*Would you want to know any information about your genetic sequence other than the specific genetic variant under investigation?*' were largely included as warm-up questions but also helped to explore participant attitudes towards different types of genetic tests. Field notes were taken by Dr. Robyn Wootton however were not included in analysis as they weren't considered to add anything beyond the transcript.

Both focus groups were audio-recorded and transcribed by an external service (described in 3.3.2). The accuracy of the transcripts was checked by replaying the audio recordings whilst simultaneously reading the transcripts. Necessary amendments were made. Transcripts were not returned to participants for notes.

3.4 Data analysis

Data analysis began after receipt of both transcripts. The transcripts were read multiple times to ensure data immersion. I used the General Inductive Approach to analyse the data: frequently arising topics were identified and transcripts were coded accordingly. These topics were then combined to form broader topics (Thomas, 2003). See Appendix 7 for coding tree. Topics were coded as they emerged from the data. Both transcripts were coded by the lead researcher, and individual quotes were reorganised in a separate documentation to form a list of quotes under each topic heading. To improve the rigour of the analysis, final topics were discussed and agreed upon with an external researcher. Participants were not given the opportunity to respond to the results.

3.5 Reflexivity

3.5.1 Personal characteristics

Both focus groups were conducted in person, thus participants knew that both the lead researcher and Dr Robyn Wootton were white, able-bodied women in their twenties. Participants were also conscious that we were researchers at the University of Bristol. This had a noticeable effect on the dynamics of the focus groups, particularly the group with an all-student sample. Participants asked multiple questions about genes and how they work in the body and about the current state and future of genomic medicine. We chose to answer questions to the best of our ability in order to allow participants to give their opinion from an informed point of view. However, on reflection, this led to an expert-learner dynamic between the researchers and participants. Whilst it was important to us that the participants felt informed about genomic medicine, future research could take steps to ensure that such a dynamic would not be repeated. For example, the presentation slide with the text '*Any questions?',* could be changed to '*Any comments?*'

Further, the lead researcher is interested in aiding the development of genomic medicine. The position taken by the lead researcher is that genomics has enormous potential for medical advancement. Indeed, whilst concerns regarding issues such as privacy are not unfounded and, of course, there is always the possibility of illegal access to and use of genetic data, in order to realise the enormous potential of genomics and to enable the correct interpretation of new genomic data, it is of central importance to be able to link patients' phenotypic data with their genomic data. This requires individuals to give consent for their data to be shared. Thus, whilst

care was taken to answer participants' questions in an unbiased way, it is possible that the lead researcher's personal views were apparent.

3.5.2 Relationship with participants

Two of the participants in the first focus group were friends of the lead researcher, and every participant in this group was a student of a similar age to the lead researcher (22 years). In contrast, there was a broader range in age in the second focus group (26 – 66 years) and none of these participants were students or previously known to the researcher. The differences between the groups' ages, student status, and relationship with the lead researcher may have influenced the dynamics between the researcher and participants. Also, it was noticeable that some participants spoke more than others. Overall, however, a good rapport between researchers and participants was created. Every participant spoke at least once.

3.6 Sample description

Sixteen participants (2 x groups of 8) were enrolled in the study (*N* =16). However, one participant in the first focus group did not attend (*N* =15). All participants had responded to a poster advert. Table 5 details the characteristics of this sample.

Table 5. Participant Characteristics in Qualitative Study.

Participant characteristics	**Focus group 1**		**Focus group 2**	
	Male n = 4	Female n = 3	Male n = 3	Female n = 5
Age				
In years	21, 18, 23, 22	23, 22, 20	27, 41, 33	26, 60, 28, 66
Data not available	0	0	0	1
Highest level of school completed				
No schooling completed	0	0	0	0
GCSEs	0	0	0	1
A Level or equivalent	3	0		0

Trade/technical/vocational training or equivalent	0	0	2	0
Bachelor's degree or equivalent	0	1	1	3
Master's degree or equivalent	1	2	0	1
Professional training/Grad scheme or equivalent	0	0	0	0
Doctorate or equivalent	0	0	0	0
Ethnicity				
White/White British	3	1	3	4
Asian/Asian British	1	0	0	1
Biracial/Biracial British	0	1	0	0
Black/African/Caribbean/Black British	0	0	0	0
Other ethnic group	0	1	0	0
Parent?				
Yes	0	0	0	2
No	4	3	3	3
Works for the NHS?				
Yes	0	1	0	0
No	4	2	3	5
Last taught about genetics				
Still studying genetics	0	0	1	0
1 – 5 years ago	3	2	0	0
5 – 10 years ago	1	1	1	1
10 – 20 years ago	0	0	1	0
20+ years ago	0	0	0	0
Never taught	0	0	0	4
Genetics in workplace environment?				
Yes	1	1	1	0
No	3	2	2	5

3.7 Results

Six topics were identified from the transcripts, including: 'detrimental psychological impact of genetic results', 'disclosure and discrimination', 'family planning', 'knowledge is power', 'genetic exceptionalism: "DNA is different"' and 'charting possible futures'. Below I summarise

each theme and provide quotes that are representative of the discussions relating to these themes.

3.7.1 Detrimental psychological impact of genetic results

Participants discussed the potential for their genetic test results to cause them "*anxiety and frustration"* (ID=F2; Female; Focus group 1). This was in reference to both results for a monogenic disorder, e.g. Huntington's, and for illnesses that can be caused by high penetrance mutations or multiple genetic variants of small effect. Due to the potential for anxiety-inducing results, some participants felt that it would be better for your mental health to *not* know your genetic risk for disease, "*ignorance is happiness as well*" (ID=F3; Female; Focus group 1).

> *… some of it [genetic results] might be quite overwhelming I guess (…) if you could get cancer or Alzheimer's or dementia, all those types of things. (ID=M1; Male; Focus group 2)*

There was a sense that a genetic diagnosis may loom over you, particularly if the test was for a late-onset disease such as Alzheimer's. Participants felt that the diagnosis could impede upon your enjoyment of life in the present moment, as it could induce stress and be "*forty years of like… thinking about, 'Oh, it's going to happen.'"* (ID=F5; Female; Focus group 2). Indeed, one participant described such a diagnosis as "*a death sentence"* (ID=F3; Female; Focus group 2).

> *I reckon I would exaggerate, even if they said you've got an extra 5% to 10% chance of something, I'd be like, "Oh, Jesus" I'd be like, "Oh, God, I'm going to get it, aren't I?" That what I feel like. So, I think I'd stress myself. (ID=M2; Male; Focus group 1)*

The possibility of an unfavourable reaction to genetic results was highlighted at a different angle; one woman touched on how the state of your mental health may influence how well you may be able to handle the information in the first place, rather than in response to the results.

I think it [effect of receiving genetic test results] depends on your mental health and if you are capable of knowing the information or not. (ID=F3; Female; Focus group 1)

One man felt that there was a lack of evidence regarding the effect that genetic results may have on your mental health and thus lessened his motivation to take part as he did not want to take the risk.

You don't really know the psychological side at all really and the effects that it is going to have. So, I would definitely hold off, (…) maybe if it was like a hundred years after or fifty years after it had already been released and stuff and there was data to suggest that it did or didn't do certain things through psychology and stuff, then maybe it would be a different case. But because you're the kind of first wave, you may be even second wave it's like… you don't really want to take that risk for yourself. (ID=M2; Male; Focus group 1)

A topic that came up frequently was the power of the mind after being told your diagnosis. Participants spoke about how anxiety from genetic results could manifest itself as hypochondria, whereby they may interpret standard sensations and bodily functions as signs of the illness for which they were (hypothetically) at genetic risk. Others discussed how the knowledge of your genetic risk may make the illness itself worse, as you may be "*waiting for it to happen"* (ID=F3; Female; Focus group 2) or that it may cause "*some kind of internal stress"* (ID=M3; Male; Focus group 1).

Do you reckon it could affect you mentally if you knew you had a high chance of something really bad, you could get really anxious about it and stuff and turn into a hypochondriac? (ID=M2; Male; Focus group 1)

(…) but then it's an odd thing because my older daughter was a nurse and she said… and I've had friends that have been told, "You've got six months to live" and almost to the day they've died. And my older daughter was saying, "The problem is, you tell somebody that and it's almost that they start dying almost, waiting for it to happen." (ID=F3; Female; Focus group 2)

Some participants were concerned about the legacy their results would have on family members. Indeed, one participant considered the possibility that knowledge of your genetic risk may induce anxiety in your children about diseases they may develop.

…if you were passing on your genome… sorry, your data to your offspring is it if you have a… a disease that they could potentially get, is that going to cause undue stress on them going throughout their whole life. (ID=M2; Male; Focus group 2)

…I would want to know for myself but if it could affect other people that I loved then maybe I wouldn't want to know. (ID=M3; Male; Focus group 2)

I don't want to mess about, if I'm having a child, I'm not sure if I should be knowing these things. But at the same time, I could be condemning that kid to a life of hell, if you know what I mean? With their future development… It would be quite interesting to know but at the same time, it would be quite… "Oh, fuck…" Sorry… it might be overwhelming for some people … (ID=M1; Male; Focus group 2)

3.7.2 Disclosure & discrimination

A common thread throughout participants' discussions was their concerns regarding the sharing of their genetic data with parties outside of the healthcare system. For example, participants discussed the possibility for their genetic data to be given to the police, and concerns about whether insurance companies may have access to the information was a particular worry for several of the participants. Another common thread that arose was the possibility of discriminatory behaviour arising from some people having "*a really good genome*" (ID=M2; Male; Focus group 1), with the implication being that another individual could have a 'really bad' genome. Indeed, one woman was concerned that *"decisions might be made because of people's genetics"* (ID=F3; Female; Focus group 2). Participants also discussed the potential for genetic data to be used in a political regime, with one woman touching on the potential for a eugenics-like, "*Hitler-y"* movement (ID=F4; Female; Focus group 2). Whilst most participants perceived the possibility of discriminatory behaviour to be undesirable, one participant suggested that it was not that big of a concern, as it would be "*just another thing that happens in society*" (ID=M3; Male; Focus group 2).

…[I am] worried about the storage of information and if it's used for insurance and employers or the police, supposing somebody makes a mistake or… (…) so hoping that it wouldn't be used intrusively. (ID=F1; Female; Focus group 2)

… it [decision to share genetic info] depends where the information goes. If it's solely kept within the NHS or if it's shared to say, for instance, insurance companies, I think that could potentially be quite dangerous. (ID=M3; Male; Focus group 2)

(…) you never know what could happen, you don't know what kind of regime might come in… that's just one example, a regime could come into politics or something and take control… anything could happen. It's not safe forever and there

are those links [between genetic data and personal identifiers] and there is the ability, it does still exist, to be able to identify you with genetic makeup, you never know what is being done with it. It's like Edward Snowden's case of him leaking the stuff that the government was doing, spying on people that wasn't exactly legal. Stuff can still be happening even though you don't know it's happening… you just gotta be aware of those things. (ID=M1; Male; Focus group 1)

One woman was concerned about the implications of an employer having access to her genetic data. She felt that this increased the intrusiveness of an already-intrusive experience of a health screening. The participant described the possibility that she could be discriminated against even if she refused to share her genetic data, as employers could interpret that decision as an attempt to hide important information.

…I think it [my concerns] would be, yes, where it could go in terms of discrimination potentially, if it got to employers or if it became something that people would want to see in terms of employers' health screening. So, I've had one health screening of "Are you fit to work for this company?" and it's very intrusive anyway, let alone if they then potentially could say, "Would you give us access to that [genetic data]?" And if you say no… obviously, maybe they couldn't but then that would discriminate them against you, "Oh, she hasn't decided to share that information" (…) That is the type of thing I'd be worried about…like is she hiding something? (ID=F5; Female; Focus group 2)

Participants also discussed how knowledge of their genetic data may influence their status as a romantic partner. It was suggested that potential partners may discriminate against you because of your genetic make-up. For example, they may decide to not start a family with you "*if you've got certain [genetic] aspects*" (ID=F4; Female; Focus group 1). One man also brought

up the possibility that knowledge of your genetic data may influence your experience of the dating world, as you may be burdened with the decision as to whether or not to share that information with potential partners.

People might say, "Oh, what's your gene sequence like?" They might not want to have kids with you if you've got certain aspects. (ID=F4; Female; Focus group 1)

There might be a separate area [for genetic data] on [the dating app] Tinder. (ID=M2; Male; Focus group 1)

M3: (…)if you were young and you got tested and you found out you had something (…) legally and morally, do you have to tell the person that you have this and there is a chance, or do you hold it back and it's going to be on your records somewhere? It's going to be confidential, but those kinds of things come up with that being a mainstream process. (ID=M3; Male; Focus group 1)

Participants discussed the importance of anonymity and how this would influence their decision to have a genetic test, *"…if it was like all anonymous and something, maybe I'd be much more likely to get it*" (ID=M3; Male; Focus group 1). Some participants felt they were more likely to get a test if the results were anonymous. Participants also questioned the extent to which genetic data could be completely anonymised and highlighted the uncertainty of future legislation regarding the anonymisation of data.

Would you ever be able to dispose of the data if you had it done and then there was going to be some change in laws or legislature, something like that; would you be able to say you don't want it on the system anymore? You said it's anonymous,

so, would they even be able to find it? (ID=M3; Male; Focus group 1)

But since it [genetic data] is anonymised why do we even care. No one is going to blame you for anything so it would be fine. (ID=F1; Female; Focus group 1)

…that [decision to share anonymised genetic data for medical research] depends on the future, what we were saying is, at the moment, in the UK, certain things are protected but it's just in the future what can happen, rules can change. And so, by doing that, it then perhaps won't be anonymous at some point in the future. I don't know if that would make a difference, but I think it would. Somebody saying to you that if you can tell us this and it's anonymous and you say yes, but in fact, twenty-five years down the line, they decide to not be anonymous and you still have family members, children, grandchildren around. I don't know how I'd feel about that really because things have happened in the past, do you know what I mean? When things change in the future it gets very different. (ID=F3; Female; Focus group 2)

How anonymous can you really make it if it's all of your personal data? (ID=F4; Female; Focus group 1)

… in the future I do have concerns about where that [my genetic data] goes and how far it goes and what it turns into. Just like everything we were talking about like data protection and things like that… (ID=F4; Female; Focus group 2

3.7.3 Family planning

Participants discussed the potential for genetic results to have an impact on their plans to have children in the first instance. This was in terms of their risk of genetic disease, *"I'm more concerned about if I would pass something onto my child"* (ID=M1; Male; Focus group 2). Participants noted that, whilst knowledge of their genetic data could enable a more informed decision as to whether or not to start a family, remaining unaware would mean they wouldn't have the burden of making such an informed decision. Further, one man discussed how the potential for knowing your carrier status may result in unwanted pressure from your friends and family to not start a family.

> *I think I'd want to know [carrier status], probably a bit controversial sounding, I'd want to know because if in not doing so, then you're causing a lot of pain to somebody, then you're going to be a bit prepared. (ID=F1; Female; Focus group 1)*

> *I guess I would like to because I don't have children, to say I do want to have children, it would be cool to know if I have that [monogenic disorder] and I've passed that on, do you know what I mean? But at the same time, I don't want to… I don't want to know because that might affect my stance of having children. (ID=M1; Male; Focus group 2)*

> *Is it possible that you might be talking to people, your family or friends that you were going to have your carrier status tested and then, there could be a level of peer pressure put on you by other people… basically, they would probably ask, "What are you going to do if you find out this or that?" And then you*

might almost feel pressured into not having a baby if… and you might want to… there could be that? (ID=M2; Male; Focus group 1)

3.7.4 Knowledge is power

Knowledge of your genetic data was also considered to have the ability to empower you to live your life to the full, as you would *"..know where you are*" (ID=F3; Female; Focus group 2). For example, participants noted that this knowledge could enable them to appreciate the people in their life more, and to make more informed decisions regarding employment.

But then, say if you were to develop Huntington's or something which might kick in when you might actually only be active when you're like thirty or something, I don't really know the full details. Surely that would be a good thing to know because you could… this is just a random example, but you could spend your life like doing a job which you're not really enjoying that much but saving up for the future. And then you end up that you develop Huntington's after that whereas, if you knew, you might want to live your life differently? (ID=M1; Male; Focus group 1)

… my mum had Motor Neurone and so she had two years, and two years is what she had. And my dad said to me that he got so much closer to her, that he cuddled her more and cherished her because he did everything for her (…) so, I just think it [knowledge of genetic risk] might well give you the time to (…) perhaps just to make people aware that you love them because we don't go around telling people we love them. So, in one way I see that knowledge is power to change that ignorance. (ID=F3; Female; Focus group 2)

Knowledge of your genetic data was also suggested to give medical professionals more power to tailor treatments to improve outcomes. Participants also noted that the development of such tailored treatments could save money and increase the efficiency of the NHS. One man also described the idea that we could use our knowledge of the function of genes to identify and eliminate the genetic cause of a disease in an individual patient.

Yes, I think a tailored treatment would be really fantastic, that is what I'm most optimistic about is, yes… tailored treatment, if they knew they wouldn't have to go through possibly as many different things, like, "Oh, let's give them this. Oh, they're not responding… oh, let's try something else." And I think if they could think, "Oh, this person is more likely to respond to this" from the start, I think that would be really great. (ID=F5; Female; Focus group 2)

…so, if they can tweak medicines or whatever to treat certain conditions that are passed on, you know, some awful conditions that children especially have to live with. If they can somehow help those hopefully, I would feel very optimistic about genetics helping that actually. (ID=F3; Female; Focus group 2)

…[genomic medicine] would make treatment a lot cheaper and a lot more straightforward than having to guess and come up with different routes I guess, yes. (ID=F3, Female, Focus group 1)

And it would save lives as well. (ID=M2, Male, Focus group 1)

It will save money in the long run as well because you won't be wasting different treatments first, you would just know how to treat them. (ID=F2; Female; Focus group 1)

They could suppress a gene with medicine or something, if they knew you had it, then go, "Take this tablet." And you're fine, you won't get it, that would be pretty good. (ID=M2; Male; Focus group 1)

Participants described how being informed about their genetic risk for disease could enable more effective preparation for illness. With regards to breast cancer, preventative measures such as a more frequent uptake of mammograms or a double mastectomy were discussed in both focus groups. Preparation was also discussed in relation to other family members, "*I would want to be mentally prepared so that I wouldn't be a burden on them* [my children]" (ID=F5; Female; Focus group 2), avoiding illness altogether, "*I'd rather take myself to Switzerland a bit earlier and do the deed than get Alzheimer's*" (ID=F3; Female; Focus group 1), as well as the practicalities of preparing for death, "*she arranged what she wanted for her funeral, she arranged all her financial stuff*" (ID=F3; Female; Focus group 2).

But like breast cancer, you could have a mammogram more frequently than people normally usually have mammograms. So, you could prevent or catch it earlier so, if you had the… what sequence… the BR…? (ID=F3; Female; Focus group 1)

BRCA. (ID=M2; Male; Focus group 1)

BRCA, you could then find it earlier. (ID=F3; Female; Focus group 1)

With breast cancer, I've heard that… I don't know if I'm right, I've heard something along the lines of if a woman does know and they have a double mastectomy that can increase their chances if they know in advance. I've heard something like that, but I don't know if it was… (…) So, I think that is like a preventative thing would be, for me, I would want to know. (ID=F2; Female; Focus group 2)

…[I would get genetically tested] with a view with not trying to think myself into an early death or something you know, like that six months to live, but with a view to living as long as possible but also being as kind as possible so that they've [my children] not got to think about what to do with me, I suppose. (ID=F5; Female; Focus group 1)

So, that's why I say it's a difficult one because, in one way, I would like to know because there are things [you can do]. We're all going to die, aren't we? (ID=F3; Female; Focus group 2)

The really negative aspects can be, well… helped a lot if you're prepared…(ID=M1; Male; Focus group 1)

Participants also touched on the capacity for the collection and sharing of genetic data to have the potential to benefit others, i.e. the 'greater good'. There was a sense that participants were aware that the larger the database of genetic and phenotypic data, the more society can gain from the scientific findings. Indeed, one man spoke about the importance of prioritising science.

I think it would be selfish to… yes, not selfish but it's better if we collect more so that we can help more people in the long run(…) you should just give your genome… (ID=F3; Female; Focus group 2)

I would agree with (…) [getting tested for the genetic variant for] Alzheimer's and things because… especially if you had family so they would know, and you could help other people… people in the future who might have the same issue. (ID=M3; Male; Focus group 2)

…they [society] always push forward scientific things which is the right thing to do. It's about survival and giving people options and choices and trying to help them. (ID=M3; Male; Focus group 2)

3.7.5 Genetic exceptionalism: "DNA is different"

A topic that came up frequently was the notion that genetic data is unlike other forms of data. Participants felt that genetic data forms a part of your personal identity in a way that other data does not, "*it's the coding of your own identity*" (ID=M3; Male; Focus group 1). Participants also felt genetic data is more private than other types of data. This unsettling feeling contributed to concerns regarding data sharing. In addition, one woman spoke about being unable to pinpoint the exact reason as to why she felt uncomfortable with the idea of sharing her genetic data with the NHS, stating that it's "*just an intrusiveness you feel*" (ID=F3; Female; Focus group 2). Again, there was a sense that this feeling of intrusiveness was tied to the notion that your genetic make-up forms a part of your personal identity which, in this participant's eyes, would mean that "*a lot of people [would] know exactly who you are".*

I just feel like you could be… probably found because it's like your… makeup. (ID=F4; Female; Focus group 1)

You know, DNA is quite a… it is a part of us, isn't it? It's very… So, yes, but I can't tell you really, really why but I think DNA is different. (ID=F3; Female; Focus group 2)

I would be much more concerned about my genetic information [being leaked than my credit card information]. You can get back money, you can't get back your y'know, it's pretty much the personal bit of information you can have. (ID=M1; Male; Focus group 1)

Well, it's literally your DNA, your code and in essence, that is your identity so, it's almost everything. (ID=M2; Male; Focus group 1)

The question of whether we 'should', from a religious point of view, know our genetic data came up in both focus groups. This perception reflects the perceived uniqueness of genetic data, as it implies that genetic data should be considered untouchable.

Are we supposed to know [if you have a monogenic disorder]? (ID=M1; Male; Focus group 2)
Do you mean from a religious point of view? (ID=F3; Female; Focus group 2)
Yes, sort of morally or ethically. (ID=M1; Male; Focus group 2)

It's a bit like playing God I guess. (ID=F3; Female; Focus group 1)

Facilitator: What do you mean by…?

Respondent: So, you shouldn't really know… you wouldn't know about this advance in technology, but we do have it, so, I don't know… (ID=F3; Female; Focus group 1)

There are some things that are meant to be…(ID=F1; Female; Focus group 1)

3.7.6 Charting possible futures

Participants envisioned the ways in which genomic medicine may develop in the future. In particular, the potential for gene editing was discussed. Some participants noted that this could occur in more isolated events, such as the genetic modification of an individual's children. Others spoke about a more wide-spread gene-editing agenda to create a better society. Whilst some participants gave the impression that they weren't necessarily opposed to genetic modification, one participant's use of the phrase "*slippery slope*" (ID=F4; Female; Focus group 2) and the statement "*I do have concerns about where that goes and how far it goes…like gene editing"* indicated the perception that genetic modification is an undesirable consequence of genomic medicine.

I think that [testing for carrier status] definitely opens up the realm for genetically modifying your kids in the future as well. (ID=M2; Male; Focus group 2)

(…) if they were able to find out…say like the president's genome code and then when they were looking to alter things in the future they could try and make people more pragmatic? (ID=F3; Female; Focus group 1)

In addition, participants spoke about their own ideas for how best to execute the delivery of genomic medicine. For example, one woman suggested that we should have an "*opt-out service*" for the donation of genomic data and that "*you should just give your genome*" (ID=F3; Female; Focus group 2). Another participant suggested that people's genetic and phenotypic data could be collected after their death.

> *So, like how they're changing it next year so, you have to donate your organs, you don't… it's an opt-out service now, not an opt-in, so, I think it should be like that and it should be… you should just give your genome… (ID=F3; Female; Focus group 2)*

> *Maybe when you're born in the future, they'll just take your code, your sequence and then, for the rest of your life they'll just be able to know how to treat you. (ID: F3; Female; Focus group 1)*

> *I'd be interested in it [getting your genome sequenced] but probably not whilst I was alive. (…) after you're dead you could say, I'd like that to be done and then you could look through what I did with my life, my records and what I died of and if it made sense. (ID: M3; Male; Focus group 2)*

3.8 Discussion

This section outlines my interpretation of the results in the context of other literature and the strengths and limitations of our study. Findings that I believe have implications for policy are discussed in Chapter 4.

3.8.1 Summary of main findings

Participants showed an impressive awareness of both the potential benefits and drawbacks of genomic medicine. Participants displayed concerns that genetic test results could: cause anxiety and impede upon enjoyment of life in the present moment; lead to hypochondria or, worse, actually increase the severity of the illness; cause stress on their children or on their plans to have children; and lead to discriminatory behaviour from insurance companies, employers, the police, or potential romantic partners. However, participants also talked about the advantages of genomics. This included the potential for genetic test results to: empower people to live their life to the full; improve the ability of medical professionals to tailor treatments to improve outcomes; allow for physical, practical, and mental preparation for illness; and enable more informed decisions regarding whether or not to have children.

In addition, participants talked about the cruciality of remaining anonymous throughout the genetic testing process. This tied in with their conversations surrounding the unique nature of DNA; participants felt that DNA conveys more personal information than other types of data. Discussions surrounding the invasiveness of genetic testing on privacy contributed to concerns surrounding the sharing of genetic data. Whilst participants acknowledged the likely benefits for science and society, participants also talked about the potential for a eugenics-like political regime. Participants also brought up the question of whether genetic research and genomic medicine is 'playing God'.

Finally, participants also put forward their own ideas for the delivery of genomic medicine. This included the proposal of an opt-out policy whereby, similar to organ donation, you are expected to donate your genomic data unless you choose not to. Another suggestion was that we should always donate our genomic and phenotypic data after death.

3.8.2 Interpretation of findings in the context of other literature

Our results mirror research from 20+ years ago that concluded the public see genetics as a 'double edged sword' (Michie et al. 1995, pg. 250) – the participants expressed both concern and excitement about genomic medicine and its future.

Unlike previous studies (Bates et al. 2005; Stockdale, Cassell & Ford, 2018; Waters, Ball, & Gehlert, 2017), the participants were given information about both the current use and anticipated future use of genetic data in the NHS prior to the discussion. Nonetheless, participants voiced thoughts and opinions that have been found consistently throughout the literature. For example, participants' concerns about the sharing of genetic data and access to genetic data by third parties have both previously arisen in other research (Hapgood et al. 2004; Trinidad et al. 2010). Similarly, past studies have also discovered worries about genetic discrimination (Fox, 2002; Gottweis, 2002) and that genomic medicine is 'playing God' (Bates et al. 2005). Indeed,the participant's perception of the potential benefits also echoed past research. For example, as in the Wellcome Trust's (2016) research, participants felt that genomics will bring advantages to both healthcare and society.

Despite discussing concerns about genetic discrimination from employers, insurance companies, and even potential romantic partners, participants did not discuss the possibility of racial discrimination, which has been found in previous research (Goldenberg et al. 2011; Peters, Rose, & Armstrong, 2004; Suther & Kiros, 2009; Middleton et al. 2018; Zimmerman et al. 2006). This may have been because the majority of participants were white, and people of colour are more likely to display such concerns (Bates et al. 2005). The Asian, biracial, and 'other ethnic group' participants in my study may have felt outnumbered and unable to discuss such concerns. This demonstrates the need for research that provides a safe space within which people of colour can confidently explore and voice their thoughts and opinions.

3.8.3 Strengths & limitations

One of the challenges of qualitative research is the potential for the researcher's opinions to bias the interpretation of the data. A strength of this study was that two researchers, with different research backgrounds, checked the coded topics leading to more rigorous analysis and discussion before a robust consensus could be reached on the topics for inclusion. Further, to my knowledge, this is one of the first qualitative studies in the UK to provide participants with some information about genetics in the NHS prior to the discussion. This allowed us to gather thoughts and opinions that are based on factual information regarding genomics as it exists now. However, this led to an expert-learner dynamic during the first focus group, and the participants asked the facilitators several questions. On reflection, this stalled the discussion and meant that there was less data on their personal opinion. Thus, a slight

alteration to the lead researcher's introduction was made for the second focus group - the lead researcher stressed that, whilst the researchers could attempt to answer questions, the field of genomics is so new that it is unlikely they would know the full answer, and that they were far more interested in hearing the participants' personal opinion.

A weakness of this study was the recruitment method. The areas in which posters were put up were generally more affluent and more likely to attract the student population. Therefore, it is likely that the sample was biased by socio-economic status (SES) and education. Whilst we did not directly measure SES, education partly makes up the social class scale and a large majority of participants had at least a bachelor's degree. In fact, all of the participants had completed education to GCSE level or above, whereas 27% of the UK population have no formal qualifications (Office for National Statistics, 2011). Whilst it is near impossible to obtain a representative sample in qualitative research of this type, and such detailed research is still a useful method to gauge the public's views and guide future research, more effort to recruit participants from a range of backgrounds was needed in this study. Indeed, we are likely to have missed important perspectives from those with working class backgrounds and/or lower education levels. Therefore, future work should aim to recruit in a diverse range of areas that ensures as much as possible that people from all backgrounds are represented in research.

On further reflection, another weakness was that the two groups of participants differed significantly from each other – one was a group of students, aged between 18–23 years, whilst the other group were all non-students and aged between 26–66 years. I originally made the decision to keep the two groups as they were because I thought younger students would feel more comfortable discussing potentially controversial opinions with their peers. On reflection, mixing the groups may have enabled a more varied discussion between the participants. The all-student group in particular may have benefited from this, as there was a tendency in this group to agree with one-another. This may have been due to a lack of confidence to challenge and contradict the status-quo amongst a group of similarly aged, younger peers. Further, two of the participants in first focus group were friends of mine. Whilst the impact of this is difficult to assess, our relationship is likely to have influenced their behaviour in the focus groups, which may have also impacted other participants' perception of the group dynamic and in turn their responses. Future work should, whenever possible, recruit only people who are unknown to the researchers involved in the study to improve scientific integrity.

Further, given the monetary reimbursement, it is difficult to ascertain whether participants' motivations were money or interest in genetics. However, due to the older age group in the second focus group (and thus higher likelihood to be employed) and the small amount offered (£10), it is plausible that these participants were more motivated by interest in genetics than by money. Again, these differences in motivation provide another reason as to why mixing the groups may have facilitated more varied discussions and may in fact be another reason why the younger participants in the first group were less motivated to disagree and have a varied discussion.

3.8.4 Conclusions

Overall, participants displayed nuanced understandings of the potential pros and cons of genomic medicine. This provides up-to-date evidence that members of the UK public see the use of genetic data in the NHS as delivering both great potential and great risk. This indicates that support for genomic medicine lies on a continuum - it is not as black and white as an individual being entirely pro or entirely anti genomic medicine. It is important that we use this information to address people's concerns and to provide a realistic picture of the benefits of genomic medicine.

3.9 Chapter summary

This chapter explored the findings of my two focus groups, whereby participants were given information about genomic medicine and asked for their thoughts and opinions. My findings were similar to past research – participants discussed concerns such as discrimination and the detrimental psychological impact of results. They also showed an awareness that genomics has many potential benefits, such as enabling more accurate diagnoses and more effective preventative measures. Considerations for the NHS and policy are discussed in the next chapter.

Chapter 4: Discussion – policy recommendations & considerations for the NHS

4.1 Chapter overview

This chapter discusses the specific policy implications that have arisen from each study, as well as broader suggestions for the integration of genomic medicine into the NHS that have become apparent through the development of my thesis. This chapter concludes with a personal reflection on the future of genomic medicine.

4.2 Policy recommendations: does knowledge of genetics predict acceptance of genomic medicine?

The first study in this thesis provided evidence against the traditional view of the deficit model (Sturgis & Allum, 2004) and offered a different interpretation of the importance of knowledge. I found that, independent of people's actual knowledge of genetics, their perception of their knowledge was the most important predictor of their acceptance of genomic medicine. In other words, for the successful development and integration of genomic medicine into UK society, my findings suggest attempts to encourage confidence in science could be included as part of the broader initiative to improve public understanding and engagement with science through education. Importantly, this confidence must not come at the cost of their knowledge; it is vital to incorporate high quality education about genetics into school science. We can encourage public confidence through accurate and clear information and collaboration with the public. Also, I should further clarify that the objective of this work is *not* to improve attitudes to genomics at all cost, but rather to encourage both engagement with and understanding of genetics and genomic medicine by providing balanced and accurate information. Indeed, it is better that the public have concerns based on fact than positive attitudes based on misinformation and/or misunderstanding. My recommendations for policy are split into two categories below – the broader implications for communication with the public (section 4.2.1)

and a more specific policy recommendation for improving public confidence in genetic knowledge (section 4.2.2).

4.2.1 Broader implications for communication with the public: inclusion of initiatives to foster scientific engagement

In addition to the passive dissemination of information to improve understanding of science, we could increase public participation in science initiatives and create and maintain an open dialogue with the public to foster more engagement with science. Indeed, given the interrelatedness of scientific understanding and scientific engagement, the following suggestions are aimed at developing successful strategies that improve upon both. For example, the educational charity 'We The Curious' focuses on removing boundaries between science and people (We The Curious, 2019). It allows visitors to 'interact with exhibits and take part in experiments' and is in the process of redesigning its venue based on questions from the people of Bristol on what makes them curious about the world. This style of approach has been found to increase the trustworthiness of researchers (Aitken, Cunningham-Burley & Pagliari, 2016) and promote a harmonious collaboration between policy-makers and the public in decision-making processes (Rowe & Frewer, 2005). Similarly, I recommend scientists attend more public events and make genetics a more mainstream public engagement activity. Showing that genetics can be part of everyday life and that it is not confined to lab experiments and clinical settings should also help to promote trust in researchers, as well as increase knowledge and confidence in genetics.

It would also be beneficial to improve our understanding of the social context in which genomic medicine will be delivered (Macintyre, 1995). This is important because the public reaction to genomics is as likely to be affected by the social environment in which the news is received as it is the perceived implications of genomics itself (Frewer, Howard & Shepherd, 1995). Thus, as well as developing an understanding of the public's confidence in their knowledge of science, we could improve our scientific understanding of the public. This includes understanding the public's opinion, decision-making processes, and personal experience, as well as a thorough investigation of the pre-existing influence of social and cultural practices and institutions, socio-economic status and the media. For example, Frewer, Howard and Shepherd (1995) identified the cruciality of credible and trustworthy information

sources in shaping positive public reaction. Future research could explore the perceived trustworthiness of organisations such as Genomics England, UK Biobank and the NHS. This could be done by asking people who visit their websites to report their level of trust in that organisation on a given scale.

4.2.2 Specific policy recommendations: how can we increase public confidence in genetic knowledge?

4.2.2.1 Promotion of successful and positive experiences with genetics and genomic medicine

Research has demonstrated that one of the most powerful approaches to improving confidence with new technology is the acquirement of personal experiences that are successful (Ertmer & Ottenbreit-Leftwich, 2010). Therefore, we suggest that science initiatives should encourage the public to apply genetic knowledge in a way that is both useful and correct. For example, they could highlight that anyone who has looked at their family history of disease has successfully applied the rules of genetics. Future research could include an experimental study to explore the effect of acknowledging such a positive experience on the acceptance of genomic medicine.

Similarly, another study discovered that confidence in technology also increases when people observe how that technology enables others' success (Ottenbreit-Leftwich, 2007). Accordingly, we propose that we should promote discussions about instances where genomic medicine has had a positive impact to the NHS, society, or science. For example, 'success stories' that reflect on a specific individual experience, scientific discovery, or public benefit. In fact, research shows that genetic technology is more accepted if it is seen to be useful (Gaskell et al. 2000), necessary (Frewer, Howard & Shepherd, 1995), or for a specific purpose (Harlander & Roller, 2012). Therefore, we recommend that genomic 'success stories' could draw upon the times in which the experience with genomic medicine showcased these three characteristics. However, we do not want to lure people into a false sense of security. All 'success stories' should give a balanced account of the ups *and* the downs, to ensure that the public have an informed understanding and realistic expectations of genomic medicine.

4.2.2.4 Summary of policy recommendations: does knowledge of genetics predict acceptance of genomic medicine?

In summary, attempts to increase the public's confidence in their genetic knowledge could be included as part of the larger initiative that aims to improve public scientific understanding. Generally speaking, this could include focussing on public engagement activities that break down the barriers between science and the public. More specifically, I recommend increased communication of 'success stories' in genetics. However, these initiatives should not come at the cost of knowledge. We can improve both confidence in knowledge and actual knowledge through increased communication of accurate and reliable information in a way that increases engagement with, as well as understanding of, science.

4.3 Policy recommendations: qualitative study – an exploration of an informed public's opinion of genetics in healthcare

Our qualitative study provided evidence that members of the UK public, after being informed about the history, current state, and future hopes of genomic medicine, see genomics as representing both great potential and great risk. In this section I have focussed on findings that hold implications for policy and the NHS.

4.3.1 Third party access & the importance of anonymity

Participants discussed the importance of maintaining anonymity and shared their concerns about third party access to their data. This finding is reflected in my quantitative study, whereby participants displayed the highest levels of concern about who would have access to their data once it is stored in an NHS database (section 2.4.6.5.3). This finding is not new - previous research has found similar concerns multiple times (Howe et al. 2018; Majumder, Cook-Deegan & McGuire, 2016; Open Data Institute, 2018). Therefore, perhaps these concerns have not been addressed sufficiently so far – we should be doing more to address them if people continue to be concerned. For example, UK residents should be given more detailed information about *how* exactly genetic data is anonymised, and *which* organisations will be allowed to access it. Information regarding the security of genetic data under GDPR, the Data Protection Act (2018) and the Cyber Security Programme in the NHS, as discussed in section

1.7.2.5, could be made more readily available to the public. However, as previously discussed, this information should be communicated in an accurate and unbiased way, with the aim to inform and not persuade. Concerns regarding illegal access to and use of data are legitimate fears and information of this type is unlikely to address or reduce them. Therefore, public concerns should be listened to in a way that offers relevant and useful information regarding legislation when necessary but does not lure people into a false sense of security when concerns are outside of the control of such legislation. Further, future research would need to explore how we can strike a balance between not overloading patients with information but giving them enough information so that they can make an informed decision. This may involve having extra information readily available for those who want more detail about data storage and security.

4.3.2 Genetic exceptionalism

Interestingly, unlike my quantitative results that revealed similar feelings of comfort towards different types of data being stored by the NHS, my qualitative study provided evidence of 'genetic exceptionalism'. Participants felt that DNA was different as it has deeper links with personal identity. My exploration of genetic exceptionalism in the quantitative study was limited to three questions that compared feelings towards genetic, mental and sexual health data and may have been confounded by the high level of trust people have in the NHS (Ipsos MORI, 2013). Contrastingly, the qualitative study did not ask for the same comparisons and allowed for a more open discussion. This difference in the conclusions between the two studies also demonstrates that qualitative research can offer a deeper insight into certain phenomena, particularly complex areas such as genomic medicine. This finding also offers some insight into the mechanism behind genetic exceptionalism – the idea that your genome is *you* in ways that other data is not, opens up the possibility for unwanted identification and thus more personally harmful misuse of this kind of data. This reinforces the importance that people place on anonymity as mentioned above. We should aim to be transparent about how genomic data is de-identified, as well as continuing to employ strict rules on who gets access to that data. Overall, it is clear that people want privacy but, given the difficulties associated with completely anonymising genetic data and the ever-present possibility of illegal access to data, the delivery of total privacy in a way that satisfies the public's wants is unlikely to be realised.

4.3.3 The importance of genetic counsellors

Participants also displayed particular concern for the impact of genetic test results on their mental health. Not only were participants concerned that genetic tests could cause them anxiety, but they talked about the potential for the knowledge of your genetic results to actually make the illness worse This highlights the importance of genetic counsellors, and again ties in with the proposal from my quantitative study that suggested both the existence and role of genetic counsellors should be emphasised in our discussions with the public (section 4.2.2.3). Genetic counsellors provide advice and support both before and after getting a genetic test result, but nearly half of the participants (43.7%) believed that meetings with genetic counsellors are only for those who have discovered they have a genetic predisposition for disease. Therefore, we should emphasise that these meetings can occur at any time throughout the genetic testing process, including *before* the patient has even decided whether or not to get a genetic test. Knowing that genetic counsellors are available to help with the decision may help to ease the anxieties regarding the impact on mental health, as well as hopefully encouraging patients to also seek expert advice and information from a genetic counsellor if required.

However, the NHS are planning to mainstream genomics more and more in the coming years, and non-genetic specialist doctors will therefore be expected to order, interpret and deliver genetic results. This may result in fewer patients having discussions with genetic counsellors. My results suggest that perhaps there should be a concerted effort to recruit more genetic counsellors, so that non-genetic specialist doctors could run their decisions past a genetic counsellor before delivering them to a patient. Alternatively, some genetic counsellors could be employed for the specific purpose of interpreting results before they are sent to clinicians. Results that are complex, or likely to require extensive emotional or practical support, could be flagged up by genetic counsellors to ensure the highest level of patient care. Of course, recruiting and training more genetic counsellors would be expensive. Whether this is viable in our cash-and-time-poor NHS is difficult however, given the consequences of misinterpreting genetic results, the NHS should give such suggestions for further investment thorough consideration.

4.3.4 The public are keen to get involved with the development of genomic policy

Participants put forward their own proposals for how the future of genomic medicine could look. For example, one participant suggested an opt-out method regarding the donation of genomic data to research. This method has been posited to decrease the burden of obtaining voluntary participation from large numbers of patients whilst allowing these patients to exercise their right to not participate should they wish to do so (Brothers et al. 2013). Research has explored the effectiveness of such an opt-out system when collecting blood samples for clinical use that would otherwise be discarded (Roden et al. 2008). This included linking the patients' genomic data to their electronic medical records. The researchers discovered that this approach can generate larger datasets with increased diversity of phenotypes. So, this suggestion from one of the participants indicates the public are not only keen to discuss ideas for the development of genomic policy, but that the ideas they put forward can be insightful and have great potential. Therefore, we could increase the number of discussions policy-makers and researchers have with the public. An excellent example of such a discussion was the recent 'Public dialogue on genomic medicine' (Ipsos MORI, 2019), that explored the public's opinion on how the NHS should mainstream genomics. This was a discussion between the public and people who work for various organisations such as Genomics England, NHS, Wellcome Trust, Department of Health and Social Care, and researchers from UK universities. This research, as well as the research in this thesis, is of critical importance for the NHS and UK Government because it expresses the opinions of the public and could be used to inform future policy.

4.3.5 Knowledge is power

Interestingly, there were wide discussions about the potential for genomics to enable a more fulfilling life in which you treasure your relationships and make more informed decisions regarding your health. Whilst this positive approach may be effective for some people, it is vital that we ensure that the public and patients are aware of the current limitations of genetic testing. Genetic results may, in fact, not reveal anything of use due to low prediction value for complex disease and our limited knowledge of genomics. People must be given accurate information that ensures they have a realistic understanding of the limits of our genetic knowledge.

4.3.6 Summary of policy recommendations: qualitative study – an exploration of an informed public's opinion of genetics in healthcare

To summarise, there should be increased communication about the current rules and legislation surrounding the security and storage of genetic data; we need to be transparent about how genetic data is anonymised and which organisations have access to it. Further, the NHS should consider investing in the recruitment of more genetic counsellors. Also, the public display enthusiasm for discussions about genomics and have insightful ideas that should be explored with further research. Finally, when communicating with the public, it is crucial that we are clear about the limits of our genetic knowledge to avoid the development of unrealistic expectations.

4.4 Future analyses & research

Below are some specific ideas for further analyses of the current dataset in the quantitative study. Also, both studies in this thesis had broader implications for research into genomic medicine.

4.4.1 Measure of confidence in knowledge

First, it is important to recognise the limitations of the methodology and analysis in the quantitative study. Participants' confidence in genetic knowledge was based on only 4 questions. In contrast, combined knowledge was based on a total of 29 questions that measured knowledge on an array of genetic topics. Therefore, it may be that specific aspects of knowledge were indeed related to acceptance of knowledge, but that this relationship was masked by other specific aspects of knowledge that were not related to acceptance. So, breaking down the larger and more general combined knowledge measure into smaller, more specific aspects of genetic knowledge may uncover associations that had previously been masked. Indeed, a total of 13 questions were removed from the final measures of biological and clinical knowledge due to a lack of internal reliability. Breaking down the combined measure into these specific aspects of genetic knowledge, such as family relatedness and heritability,

may offer some useful insight into the lack of inter-relatedness between these items and the final items.

Another way of doing this could be to focus on the themes within the four questions in the perception of knowledge measure. These specifically asked about participants' confidence regarding their knowledge of the terms 'DNA', 'human genome', 'genetically modified', as well as their self-reported understanding of the ethical issues raised in genetic research. Thus, future analyses of this data set could include the creation of four clusters from items in the combined measure: understanding of DNA, understanding the human genome, understanding genetic modification and demonstrating an awareness of ethical issues. Multiple regression analyses using these four clusters as individual predictor variables would indicate their ability to predict acceptance of genomic medicine. Indeed, given that confidence in these four aspects of knowledge predicated acceptance of genomics, it may be that *actual* knowledge of these features of genetics is associated with acceptance. If so, perhaps we should focus on encouraging knowledge of and engagement with specific parts of the broad subject matter that is genomic medicine. It would also be interesting to explore the relationship of these new predictor variables with each corresponding confidence measure. For example, exploration of the correlation between confidence in knowledge of DNA and actual knowledge of DNA would enable a more valid conclusion as to whether the public are accurate in estimating their genetic knowledge.

4.4.2 Exploration of negative correlation between being a parent and knowledge

In the past, researchers have hypothesised that parents would have *higher* genetic knowledge than people with no children, given that they may actively seek out genetic information that may be relevant for their child/children (Chapman et al. 2019). However, this same research actually revealed *no* significant difference in knowledge between those who have children and those who do not. In the present study, there was a negative correlation between being a parent and both actual and perceived knowledge of genetics. Further analysis of this finding could include multiple regression that controls for the effects of age (age was also negatively correlated with both measures in our study) as parents are likely to be older than people with no children. Age may have confounded the association given that it is linked to the length of

time since people were in education. Also, as genetics and genomic medicine are becoming more mainstream, it is likely that there has been an increase in the amount of genetics taught at school. Therefore, older people who went to school before this surge in genetics education may have a lower level of genetic knowledge. This would be interesting to explore as it could indicate whether educational initiatives should include a concerted effort to target those of an older age.

4.4.3 Future research should include a measure of religious beliefs

The finding in my qualitative study that participants were concerned that genomics was 'playing God' served as a reminder that I should have included a measure of religious beliefs in my quantitative study. Indeed, Allum et al. (2014) found that Catholics are less supportive of genetic testing on unborn babies than those without religious beliefs, and other research has found that atheist participants displayed higher genetic knowledge than their Christian counterparts (Chapman et al. 2017). Further, Allum et al. (2014) found that religion can act as a 'perceptual filter' and affect the way in which scientific knowledge affects attitudes to genetic testing, i.e. despite their finding that more knowledgeable participants were more supportive of genetic testing, this relationship was reversed if the participants were also highly religious. This finding is particularly relevant to my quantitative study because the potential presence of religious beliefs may have negatively affected the strength of the relationship between knowledge and acceptance. Future research should include a measure of religious beliefs to explore whether it has an effect on the relationship between knowledge and acceptance.

4.5 The future of genomic medicine: a personal reflection

I have spent this year reading widely about the history, current state, and future of genomic medicine in the UK. In addition, I have attended the Festival of Genomics in London and the European Society of Human Genetics conference in Gothenburg and have collaborated with the West of England Genomic Medicine Centre.

It has become clear that without the careful consideration and exploration of public opinion and concerns, we run the risk of missing the full range of opportunities to advance medical

frontiers on both a national and global level. Public trust and support is important in order to secure a successful future for genomics. However, whilst the NHS and policy-makers should continue to address the concerns that were discussed in the focus groups, this should be conducted in an unbiased way that aims to provide accurate and relevant information that helps people come to their own conclusion, i.e. the provision of this information is to inform and not persuade. Also, some concerns, such as illegal use of genetic data or accidental leaking of genetic data, are unlikely to be addressed with the provision of information regarding the law and/or legislation surrounding genetic data. The majority of concerns such as these should simply be listened to and recognised as serious anxieties.

Also, findings regarding the importance of confidence in genetic knowledge in chapter 2 should be analysed further, e.g. breaking down the combined knowledge measure to tap into more specific aspects of genetic knowledge and their influence on acceptance. Future research should include a personality measure to control for the effects of general confidence and high self-esteem, as well as recruiting a more diverse sample in terms of SES, education, and ethnicity. Indeed, the recent decision by the government to *not* charge people for voluntary genomic testing due to fears over creating a two-tiered healthcare system reflects an inclusive approach to healthcare that we should continue to uphold in research.

Overall, we are at the beginning of the implementation of genomic medicine into routine care. It is an incredibly exciting time for genomics; it has been a privilege to conduct research into this field at this time.

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Appendices

Appendix 1 Screenshot of online ethics application and approval for quantitative study.

Application Review

Logout

ID	Name	Faculty	Department	Supervisor
75841	Miss Helena Davies	Faculty of Science	Psychological Science	Professor Claire Haworth

Status
Signed off
Date added
Oct. 9, 2018
Signed off date
Oct. 12, 2018
Is this a student project?
Postgraduate Masters
Project title
An online survey to explore public knowledge of genetics and public awareness and acceptance of the use of genetics in healthcare
Estimated start date
Nov. 1, 2018
Duration (months)
12
Project outline
Research questions: This study will involve an online questionnaire that aims to explore: What is the current level of knowledge of genetics and awareness of the situation regarding the movement of genetics into healthcare in the UK public? How much do the UK public accept the movement of genetics into healthcare? Is there a link between knowledge, awareness and acceptance? Methods: Participants for this study will be recruited via social media and email. There is also the possibility for recruitment to take place at various science and non-science festivals, and at local attractions such as We The Curious. The likely sample size is 300 participants, all of which must be over 18 years of age and residents in the UK. A draft questionnaire is attached for viewing. Essentially, it includes questions that will first test participants' knowledge of genetics and awareness of the use of genetics in healthcare. The final part of the questionnaire provides information regarding the current status of genetics in healthcare, followed by questions that explore public acceptance. Importance: First, given the constantly evolving field of genetics research, it is crucial to ensure that current understanding of public opinion is up to date. Attempts to understand public attitude towards genetics could uncover gaps in knowledge, which in turn can be used to guide communication efforts (Henneman et al., 2012). Indeed, a lack of understanding may discourage involvement in medicine that incorporates genetic information (Sperber et al. 2017) and result in a rejection of genetics research at the very time that the medical usefulness of such information is increasing (Waters, Ball & Gehlert, 2017). Second, it is often assumed that anxiety towards new technologies is a result of a low level of knowledge (Jallinjoa & Aro, 2002). By this logic, improving the general public's knowledge of genetics should work to ensure a positive reaction towards the integration of genetics within healthcare. However, there is conflicting evidence regarding the link between knowledge of genetics and acceptance; Eisendel (2000) found no link, whilst another study found that more knowledgeable individuals were more optimistic about genetic tests (Allum et al. 2014). Contrastingly, Jallinoja and Aro (2002) found that knowledge was linked to both more scepticism and enthusiasm. Therefore, this study will investigate the link between the two within the UK public. Finally, many studies within this field are conducted outside of the UK (Jallinjoa & Aro, 2002; Henneman et al., 2012; Waters, Ball & Gehlert, 2017; Condit & Shen, 2011). Therefore, it is vital to consider the views of the UK public, not only because differing opinions on genetics may arise as a result of cultural sensitivities or historical influences (Gaskell et al., 1999), but also because the UK is the first country in the world to bring genetics into a mainstream healthcare system. (References attached)

1. Does the research involve human participants? If you answered No, please go to question 2.	Yes
1a. Does the research involve participants who are particularly vulnerable or unable to give informed consent? Examples of vulnerable participants or those unable to give informed consent are children, people with learning difficulties, patients, people experiencing emotional distress or mental illness, people living in care or nursing homes, and people recruited through self-help groups, participants in a dependent or unequal relationship with the researcher(s) or research supervisor.	No
1b. Will it be necessary for participants to take part without their knowledge and consent at the time? Examples include the covert observation of people	No
1c. Will the research involve actively deceiving participants? Examples include deliberately falsely informing participants, withholding information from participants or misleading participants in such a way that they are likely to object or show unease when debriefed about the study.	No
1d. Will the research involve discussion or collection of information on sensitive topics? Sensitive topics under the Data Protection Act 1998 include: ◦ The racial or ethnic origin of the data subject; ◦ Their religious beliefs or other beliefs of a similar nature; ◦ Whether they are a member of a trade union (within the meaning of the Trade Union and Labour Relations (Consolidation) Act 1992); ◦ Their physical or mental health or condition; ◦ Their sexual life; ◦ Their commission or alleged commission by them of any offence; ◦ Any proceedings for any offence committed or alleged to have been committed by them, the disposal of such proceedings or the sentence of any court in such proceedings. If the research is in relation to any of the sensitive topics listed under the DPA 1998 then the legal issue requiring such scrutiny in such cases that 'explicit consent' must be obtained.	No
L1 - Does your research involve any of the following? ◦ Medical devices, ionising radiation, drugs, placebos or other substances to be administered to participants. ◦ Adults (over 16) who lack capacity to consent for themselves including participants, who will be retained in the study following loss of capacity. ◦ Recruiting or using client data from NHS patients, nursing home/independent hospital/clinic or medical agency patients, users of social care services or prisoners. For more details on definitions please see 'Does my project require NHS review': http://www.nres.nhs.uk/applications/approval-requirements/ethical-review-requirements/requirements-for ethical-review-under-legislation/ (this link opens in a new window).	No
L2 - Does your research involve any of the following: ◦ Human Blood or Tissue Samples (Tissue means any relevant material consisting of or including cells - for definition of 'relevant material', please see the Human Tissue Authority website at http://www.hta.gov.uk/ - this link opens in a new window.	No
L3 - Does your research involve any of the following ◦ Animals (either use or observation)	No
L4 - Does your research involve any of the following? ◦ Has or will your research be submitted to another ethics committee?	No
L5 - Does your research involve any of the following? ◦ Working or travelling overseas	No
L6 - Does your research involve any of the following? ◦ Trials outside the UK ◦ Pregnant research subjects ◦ Conception/Contraception ◦ Children under 5 ◦ More than 1500 research subjects ◦ Genetic engineering ◦ Hepatitis/CJD/HIV & Aids related research	No

1e. Does the research involve invasive procedures? Invasive procedures may include: ◦ Administration of drugs placebos, or other substances (e.g., drinks, foods, food or drink constituents, dietary supplements) to study participants; ◦ Biological samples from participants be obtained; ◦ Pain or more than mild discomfort likely to result from the study.	No
1f. Does the research involve scans or x-rays of research participants?	No
1g. Does the research involve photographs, videoing, recording or similar of research participants?	No
1h. Will financial inducement (other than reasonable expenses and compensation for time) be offered?	No
1i. Will the study involve the use or storage of information about living people whose personal identity could be discovered from that information?	No
1j. Does the study risk causing psychological stress or anxiety or other harm or negative consequences beyond that normally encountered by the participants in their life outside research?	No
2. Will the research involve politically and culturally sensitive funding sources? Examples include the defence sector, projects with potential environmental effects and other internationally regulated or protected industries. For more information, please follow the link to the 'Research Governance and Integrity Policy': http://www.bris.ac.uk/red/support/governance/RGI.pdf (this link opens in a new window).	No
3. Will the research involve politically, culturally or socially sensitive topics? For more information, please follow the link to the Faculty of Arts Ethics Committee Guidance Note (PDF 78kb) (this link opens in a new window).	No
Supporting information (up to approximately 300 words) Please provide any additional information in relation to your study such as adhering to a particular SOP or confirming if your study is a service evaluation/audit as opposed to research.	

Appendix 2 Survey questions with reference to their sources.

[Demographic information]

1. How old are you? Please indicate your age in years.

_____

2. To which gender do you identify most?

- Male
- Female
- Non-binary
- Prefer not to say

3. To which ethnic group do you identify most?

- White/White British
- Asian/Asian British
- Black/African/Caribbean/Black British
- Biracial/Biracial British
- Other ethnic group
- Prefer not to say

4. Do you have any children?

- Yes
- No
- Prefer not to say

5. What is the highest degree or level of school you have completed or are currently enrolled in?

- No schooling completed
- GCSEs or equivalent
- A Levels or equivalent
- Trade/technical/vocational training or equivalent
- Master's degree or equivalent
- Professional training/Graduate scheme or equivalent
- Doctorate degree or equivalent
- Prefer not to say

6. When were you last taught about genetics?

- Still studying genetics
- 1-5 years ago

- 5-10 years ago
- 10-20 years ago
- 20+ years ago
- Have never been taught about genetics
- Prefer not to say

7. Do you come across genetics in your workplace?
    - Yes
    - No
    - I am not in paid employment
    - Prefer not to say

[Experience with genetic testing]

1. Before beginning this survey, had you heard of genetic testing?
    - Yes
    - No
    - Prefer not to say

2. Have you or anyone close to you ever had a genetic test done by a doctor?
    - Yes
    - No
    - Prefer not to say

3. Have you ever used an online genetic test to assess your genetic health risk?
    - Yes
    - No
    - Prefer not to say

4. Have you ever used an online genetic test to assess your ancestry?
    - Yes
    - No
    - Prefer not to say

[Biological knowledge of genetics]

Below are some questions about genetics and their role within the body. Please answer these questions to the best of your ability.

1. Which of the following is TRUE:
   - It is the mother's genes that determine the sex of the child[1]
   - By eating a genetically modified fruit, a person's genes could also become modified[1]
   - Humans share approximately 99% of their DNA with each other[1]

2. Which of the following is FALSE:
   - By eating a genetically modified fruit, a person's genes could also become modified[1]
   - The cloning of living things produces genetically identical copies[1]
   - All plants and animals have DNA[1]

3. Which of the following is TRUE:
   - More than half of human genes are identical to those of mice[1]
   - An individual who is a carrier of a genetic disease will develop the disease as they age
   - Genes are always more important for how you look than how you behave

4. Which of the following is FALSE:
   - Your genetic make-up can influence the way that you respond to medical interventions[2]
   - Cancer can be caused by both inherited genetic mutations or mutations that are acquired throughout the lifetime[2]
   - A 'dominant trait' is a trait that is most popular in a single population

(5. Which of the following is FALSE: Excluded as there is more than one correct answer)
   - Environmental factors, such as cigarette smoke, can affect gene activity[3]
   - When someone says something is "epigenetic" it means that you can inherit changes in gene activity without inheriting changes in the genes[3]
   - Most of the human genome consists of genes that code for proteins[3]

6. Which of the following is TRUE:
   - Traits with higher heritability are more difficult to change[4]
   - Interventions should be targeted to traits influenced by the environment[4]
   - Environmental interventions can be used to mitigate genetic risk[4]

7. Which of the following is FALSE:
   - DNA is contained in the nucleus of the cell[4]
   - DNA stands for Deoxyribonucleic acid[4]
   - DNA uses a four letter code[4]
   - The bases of DNA are AUGC[4]

Are the following statements true or false?

8. Most physical human traits, e.g. height, are controlled by a single gene[3]
   - True
   - False

9. A gene codes directly for a trait or disease[3]
- True
- False

10. A single gene can influence several different traits or diseases[3]
- True
- False

Please answer the following questions to the best of your ability.

11. How genetically similar is a person to their identical twin's daughter?[4]
- 12.5%
- 25%
- 100%
- 33.3%
- 50%

12. The word 'heritability' means…[4]
- The proportion of the variation in the physical composition of a population accounted for by genetic variation
- The proportion of a physical composition that is passed on to the next generation
- The proportion of a person's characteristic that is accounted for by genes
- The proportion of genes that are important for the development of a characteristic

13. If you share 50% of your DNA variants with your sister, how much do you share with your cousin?[4]
- 50%
- 25%
- 12.5%
- 8%

14. What is a genome?[5]
- A sex chromosome
- The entire sequence of an individual's DNA
- All the genes in DNA
- Gene expression

15. What is the main function of all genes?[5]
- Storing information for protein synthesis
- To provide energy to the cell
- To clear out waste from the cell

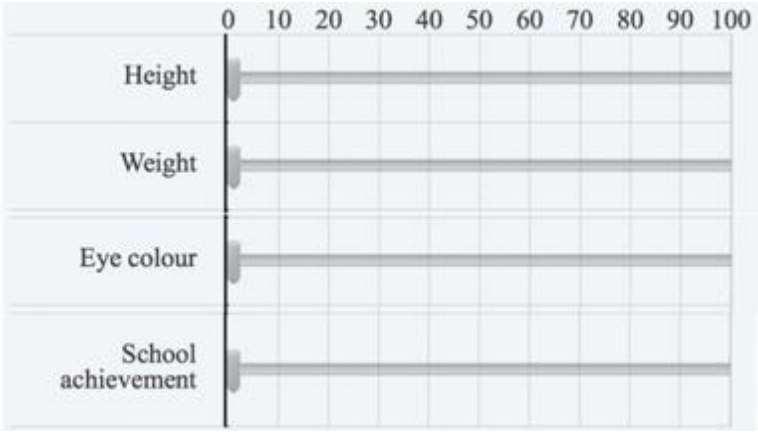
- To repair damage to a cell

16. Approximately how many genes does the human DNA code contain?5
- 2,000
- 1 million
- 3 billion
- 20,000

17. The DNA sequence in two different cells, for example a neuron and a heart cell, of one person, is:5
- Entirely different
- About 50% the same
- More than 90% the same
- 100% identical

18. -Non-coding DNA describes DNA that:5
- Is removed when passed from parent to offspring
- Does not lead to the production of proteins
- Is non-human DNA
- Is not composed of nucleotides

19 - 22. On a scale of 0-100 how important do you think genetic differences are between people in explaining individual differences in the following traits (with 100 = only genetic differences can explain individual differences in traits):5



Please read the following scenarios and select the answer you believe to be correct.

23. A doctor tells a couple that they've got a one in four chance of having a child with genetic disease...6
- ...if their first three children are healthy, the fourth will have the illness

- ...if their first child has the illness, the next three will not
- ...each of the couple's children will have the same risk of suffering from the illness
- ...if they have only three children, none will have the illness

24. A patient is told that the results of their genetic test indicate they are at increased risk of a particular disease. Consequently, they decide to engage in healthier behaviours, such as exercising regularly and eating a more balanced diet.

- ...their increase in healthier behaviour will have no effect on their likelihood of developing the disease, because their genetic test indicates they will definitely develop the disease at some point
- ...their increase in healthier behaviour will definitely decrease their likelihood of developing the disease, because the effect of the environment will always override the influence of their genes
- ...their increase in healthier behaviour may lead to a decrease in their likelihood of developing the disease, because the way their genes work can be altered by their environment

25. The Smith family consists of a mother, father, a pair of identical twins, and a daughter.

- ...the parents each share 50% of their DNA with each of their children, the identical twins share 100% of their DNA with each other, and the daughter shares 50% of her DNA with each member of the family
- ...the parents each share 50% of their DNA with each of their children, the identical twins share 50% of their DNA with each other, and the daughter shares 50% of her DNA with each member of the family
- ...that the parents each share 50% of their DNA with each of their children, the identical twins share 100% of their DNA with each other, and the daughter shares 100% of her DNA with each member of the family
- ...the parents each share 100% of their DNA with each of their children, the identical twins share 100% of their DNA with each other, and the daughter shares 50% of her DNA with each of the twins

[Perception of genetic knowledge]

1. When you hear the term DNA, how would you rate your understanding of what the term means? 1

- Very good
- Good
- Some understanding
- Heard the term, but little understanding of what it means
- Have not heard the term
- Don’t know

2. When you hear the term ‘human genome’, how would you rate your understanding of what the term means? 2

- Very good
- Good
- Some understanding
- Heard the term, but little understanding of what it means
- Have not heard the term
- Don’t know

3. When you hear the term GM or ‘genetically modified’, how would you rate your understanding of what the term means?3

- Very good
- Good
- Some understanding
- Heard the term, but little understanding of what it means
- Have not heard the term
- Don’t know

4. Please indicate how much you agree or disagree with the following statement…I feel I have a good understanding of the ethical issues raised by genetic research4

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

[Clinical knowledge of genetics in healthcare]

Before you begin the next section, please bear in mind that the phrase 'genetics in healthcare' refers to the use of an individual's genetic information to inform their clinical care.

This next section includes statements about the current state of genetics in UK healthcare. Please indicate whether you believe them to be true or false.

1. Using an individuals’ genetic code, we are currently able to predict whether an individual will develop every single disease known to the medical community

- True

- False

2 There are many common diseases where the study of genetics can show the road to better treatment[1]

- True
- False

3. The use of genetics in healthcare can mean that expensive drugs are only given to those who will benefit from them[1]

- True
- False

4. It is against the law for a doctor to **not** disclose a patient's genetic test results to their close relatives as, given their genetic relatedness, the results may also concern them[2]

- True
- False

5. Currently, treatment for cancer can include a genetic diagnosis to show if a tumour might respond to a certain treatment[3]

- True
- False

6. The use of patients' genetic data to deliver targeted therapies is already changing people's lives[1]

- True
- False

7. A patient will only speak with a genetic counsellor if their genetic test result indicates they have a genetic predisposition to a disease[4]

- True
- False

8. Through a number of genetic testing techniques, it is possible to detect genetic abnormalities in an unborn child[5]

- True
- False

9. The rapidly expanding role of genetics in many healthcare decisions is already increasing the demand for qualified genetics professionals[1]

- True
- False

10. Currently, the NHS has every individual patient's genetic code stored on their database
    - True
    - False

11. At present in the UK, newborn infants are tested for certain genetic traits[6]
    - True
    - False

12. How soon is the NHS planning to include genetics in routine healthcare?[7]
    - In the next 10 years
    - In the next 25 years
    - In the next 50 years

The next 4 questions are aimed at exploring your more general awareness of the use of genetics in healthcare. Please answer these questions as truthfully as possible.

13. How much have you read or heard about genetic tests that predict the likelihood that a person will develop certain genetically influenced diseases or conditions, such as heart disease, cancer and Alzheimer's?[8]
    - Quite a lot
    - Some
    - Not much
    - Nothing at all
    - Don't know

14. Before beginning this survey, were you aware of the use of genetics in healthcare?
    - Yes
    - No
    - Prefer not to say

15. Have you heard of the 100,000 Genomes Project?
    - Yes
    - No
    - Prefer not to say

16. Have you heard of Genomics England?
    - Yes
    - No
    - Prefer not to say

17. Have you heard of the phrase 'personalised medicine?'
    - Yes

- No
- Prefer not to say

[Acceptance of genetics in healthcare]

Before you begin this final section, please read the information below from Genomics England and the NHS regarding the current status of genetics in healthcare[1,2]. You can refer back to this information at any point when answering the next set of questions.

In 2012, the UK Government set out to gather 100,000 complete sets of genetic information (genomes) from patients with cancer or a rare disease (using a process called 'whole genome sequencing'). On the 5th December 2018 they reached their target, and now the NHS will provide a whole lifetime of medical records from each participant of the project and link it to their genetic information. This will help us to tease apart the complex relationship between our genes, what happens to us in our life, and illness.

The NHS is the first healthcare system in the world to do this on such a large scale. This information will provide a unique research database, thus enabling a powerful learning system able to provide better outcomes for patients.

Now, the UK wants to reach a target of 5 million genomes by 2023/24. To help reach this target, patients with a rare disease or cancer will now begin to be offered whole genome sequencing by the NHS as part of routine care. For these patients, this will enable more comprehensive and precise diagnosis and access to more personalised treatment. Also, healthy people in England will also be given the option to undergo whole genome sequencing, however the test will cost them (predicted to be hundreds of pounds).

Overall, the aim is to create a new genetic medicine service for the NHS - transforming the way people are cared for. For example, patients may be offered a diagnosis where there wasn't one before, or may be able to make more informed decisions regarding their treatment. And in time, there is potential for the development of new and more effective treatments. NHS England states that this approach to medicine takes greater account of people's genetic differences, rather than a "one-size-fits-all approach to the treatment and care of patients with a particular condition".

1. First, please indicate how optimistic you are about the possibility of medical advances as a result of genetic research[3]

1. Not at all optimistic
2. Not too optimistic
3. Somewhat optimistic
4. Very optimistic
5. Prefer not to say

Now, please indicate how much you agree with the following statements regarding genetics and the use of genetics in healthcare.

2. If I was told that knowledge of my genetic data may improve the effectiveness of a medical intervention that I required, I would not hesitate to have my genetic data tested

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

3. The use of genetic data to inform medical decisions is overly intrusive

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

4. I am concerned about who will have access to my genetic data once it is stored within an NHS database

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

5. The benefits that the use of genetics in healthcare will bring to our healthcare system far outweigh any potential downsides

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

6. I do not welcome the notion of genetics in healthcare because insurance companies will ask for genetic information to decide how at-risk one is for ill health, which may increase the price of health insurance

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

7. If my genetic data was included in research which eventually led to the development of a medical drug, I would not be happy for a commercial company to profit from the selling of this drug

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

8. I would be more concerned about my genetic data being leaked than my credit card information[4]

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

9. Family members share many genetic traits and may have the same genetic abnormalities associated with disease. Therefore, all immediate family members must give consent before an individual in that family gets a genetic test for medical reasons[5]

- Strongly agree
- Moderately agree
- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

10. I believe that my destiny is written in my genes[6]

- Strongly agree
- Moderately agree

- Slightly agree
- Slightly disagree
- Moderately disagree
- Strongly disagree

11. I do not trust the healthcare system in the UK because it might misuse genetic data obtained from patients[6]
    - Strongly agree
    - Moderately agree
    - Slightly agree
    - Slightly disagree
    - Moderately disagree
    - Strongly disagree

12. I feel suspicious about genetic studies for the improvement of health: hidden political/economic agendas may be behind them[6]
    - Strongly agree
    - Moderately agree
    - Slightly agree
    - Slightly disagree
    - Moderately disagree
    - Strongly disagree

13. Scientific development, including the research in genetics, is essential for improving people's lives[6]
    - Strongly agree
    - Moderately agree
    - Slightly agree
    - Slightly disagree
    - Moderately disagree
    - Strongly disagree

14. Preventing health problems is preferable to curing health problems
    - Strongly agree
    - Moderately agree
    - Slightly agree
    - Slightly disagree
    - Moderately disagree
    - Strongly disagree

The next 3 questions consider the various types of medical data that can be stored by the NHS. Currently, the NHS keeps medical records which will include keeping track of any appointments you may have had with your GP. Please indicate how much you agree with the following statements.[7]

15. I would feel uncomfortable with the possibility for the NHS to have my genetic data on record
    - Strongly agree
    - Moderately agree
    - Slightly agree
    - Slightly disagree
    - Moderately disagree
    - Strongly disagree

16. I feel uncomfortable with the possibility that the NHS have my sexual health data on record
    - Strongly agree
    - Moderately agree
    - Slightly agree
    - Slightly disagree
    - Moderately disagree
    - Strongly disagree

17. I feel uncomfortable with the possibility that the NHS have my mental health data on record
    - Strongly agree
    - Moderately agree
    - Slightly agree
    - Slightly disagree
    - Moderately disagree
    - Strongly disagree

18. Now, please indicate which statement you agree with most:[8]
    - For me, DNA information is the same as any other medical information, like blood pressure or blood sugar levels
    - For me, DNA information is different to other medical information because, for example, it tells us how we are related to other people
    - I'm not sure

19. Finally, imagine yourself in the following scenario. You have an opportunity to have a genetic test that may reveal that you are going to develop a condition that currently does not have any medical treatment options. Which statement do you agree with **most?**[9]

- **Knowledge is power**: I would rather have the genetic test, so that I can prepare financially, emotionally, and practically if I am found to have a genetic variant that will lead to an (currently) incurable disease
- **Ignorance is bliss:** I would rather not have a genetic test, because if I am found to have a genetic variant that will lead to an (currently) incurable disease I would worry about it too much to be able to carry on enjoying my life to the full
- I'm not sure

That is the end of the survey - thank you for your time.

Question sources:
Biological knowledge of genetics

1. [Adapted from] Wellcome Trust Monitor. (2016). Wave 3: Chapter 7: What does the public know about science and medical research? Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave3-full-wellcome-apr16.pdf

2. Discussion with a genetic counsellor.

3. [Adapted from] Carver, R. B., Castéra, J., Gericke, N., Evangelista, N. A. M., & El-Hani, C. N. (2017). Young adults' belief in genetic determinism, and knowledge and attitudes towards modern genetics and genomics: the PUGGS questionnaire. *PloS one*, *12*(1), e0169808.

4. [Adapted from] Haworth, C. (2018). Genes and Behaviour course assessment. University of Bristol.

5. Chapman, R., Likhanov, M., Selita, F., Zakharov, I., Smith-Woolley, E., & Kovas, Y. (2017). Genetic Literacy And Attitudes Survey (Iglas): International Population-Wide Assessment. In *The European Proceedings of Social & Behavioural Sciences EPSBS* (pp. 45-66).

6. Wellcome Trust Monitor. (2012). Wave 1. Chapter 3.5: Understanding probability in science. Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave1-wellcome-sep09.pdf?cv=1

Question sources:
Perception of genetic knowledge

1. Wellcome Trust Monitor. (2012). Wave 1. Chapter 3: Understanding of key terms in medical research. Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave1-wellcome-sep09.pdf?cv=1

2. Wellcome Trust Monitor. (2013). Wave 2: Chapter 2.5: Awareness and understanding of issues related to genetic research. Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave2-full-wellcome-may13.pdf

3. Wellcome Trust Monitor. (2016). Wave 3: Chapter 7: What does the public know about science and medical research? Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave3-full-wellcome-apr16.pdf

4. Wellcome Trust Monitor. (2012). Wave 1. Chapter 8: Knowledge and understanding of genetic research. Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave1-wellcome-sep09.pdf?cv=1

Question sources:

Clinical knowledge of genetics in healthcare

1. Genomics England. (2018). Retrieved from https://www.genomicsengland.co.uk/about-genomics-england/

2. Chico, V. (2017). ABC v St George's Healthcare NHS Trust: an arguable duty to disclose familial genetic risks. *Bionews.*

3. AACC. (2016) Genetic Tests for Targeted Cancer Therapy. Retrieved from https://labtestsonline.org/tests/genetic-tests-targeted-cancer-therapy

4. Discussion with a genetic counsellor.

5. NHS. (2018). Your pregnancy and baby guide: Screening tests in pregnancy. Retrieved from https://www.nhs.uk/conditions/pregnancy-and-baby/screening-tests-abnormality-pregnant/

6. Chapman, R., Likhanov, M., Selita, F., Zakharov, I., Smith-Woolley, E., & Kovas, Y. (2017). Genetic Literacy And Attitudes Survey (iGLAS): International Population-Wide Assessment. In *The European Proceedings of Social & Behavioural Sciences EPSBS* (pp. 45-66).

7. NHS England. (2016). IMPROVING OUTCOMES THROUGH PERSONALISED MEDICINE. Working at the cutting edge of science to improve patients' lives. Retrieved from https://www.england.nhs.uk/wp-content/uploads/2016/09/improving-outcomes-personalised-medicine.pdf

8. [Adapted from] Wellcome Trust Monitor. (2016). Wave 3: Chapter 7: What does the public know about science and medical research? Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave3-full-wellcome-apr16.pdf

Question sources:

Section 4 – Acceptance of genetics in healthcare

1. Genomics England. (2018). Retrieved from https://www.genomicsengland.co.uk/about-genomics-england/

2. NHS Genomic Medicine Service. (2017). Retrieved from https://www.england.nhs.uk/genomics/nhs-genomic-med-service/

3. [Adapted from] Wellcome Trust Monitor. (2012). Wave 1. Chapter 8.5: Optimism about genetic research. Retrieved from https://wellcome.ac.uk/sites/default/files/monitor-wave1-wellcome-sep09.pdf?cv=1

4. Chen, A. (2018). Why a DNA data breach is much worse than a credit card leak. The Verge. Retrieved from https://www.theverge.com/2018/6/6/17435166/myheritage-dna-breach-genetic-privacy-bioethics

5. Chico, V. (2017). ABC v St George's Healthcare NHS Trust: an arguable duty to disclose familial genetic risks. *Bionews.*

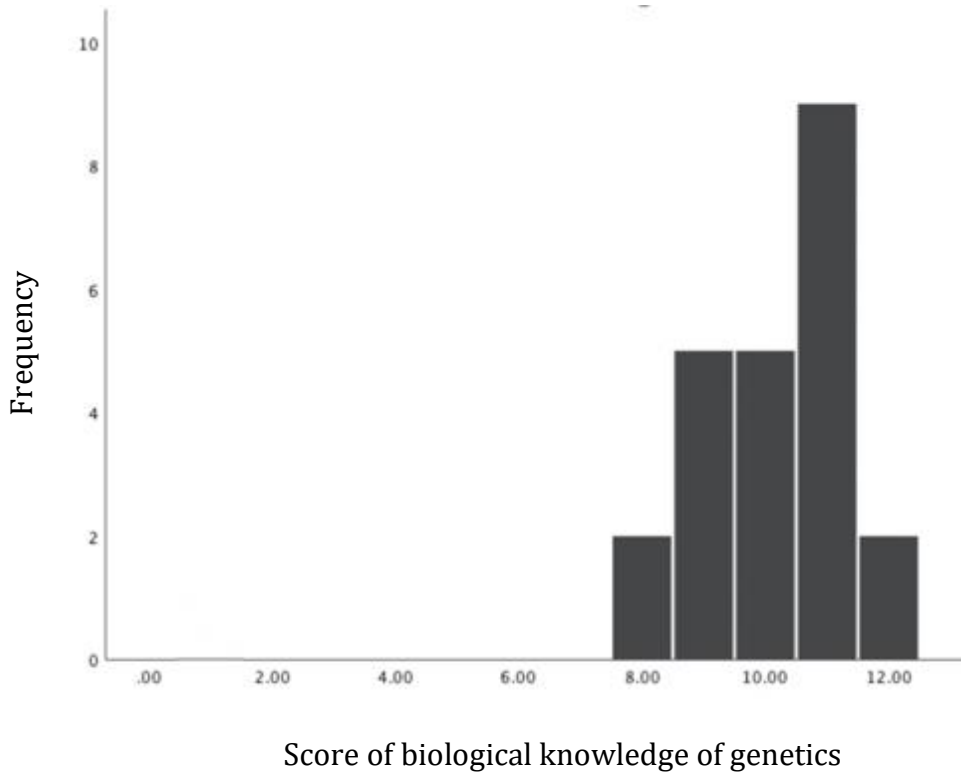
6. Chapman, R., Likhanov, M., Selita, F., Zakharov, I., Smith-Woolley, E., & Kovas, Y. (2017). Genetic Literacy And Attitudes Survey (iGLAS): International Population-Wide Assessment. In *The European Proceedings of Social & Behavioural Sciences EPSBS* (pp. 45-66).

7. 'Genetic exceptionalism' in Davies, S. C. (2017). Annual report of the chief medical officer 2016, generation genome.

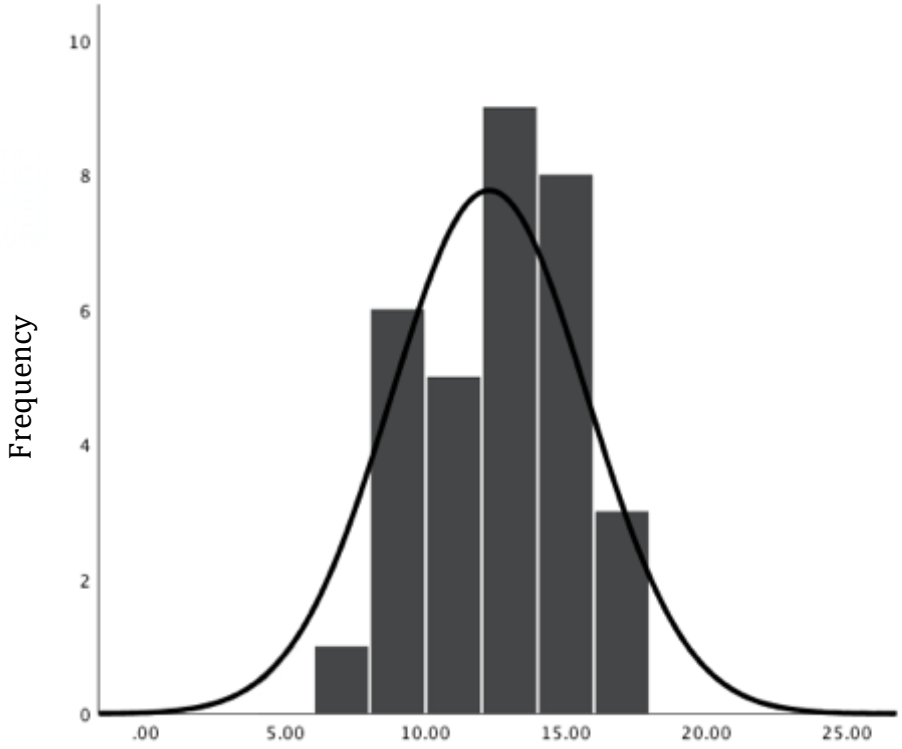
8. [Adapted from] Middleton, A. (2017). Your DNA, Your Say. *The New Bioethics*, *23*(1), 74-80.

9. Plomin, R. (2018) Blueprint: How DNA Makes Us Who We Are. Annual Lecture at University of Bristol, Bristol.

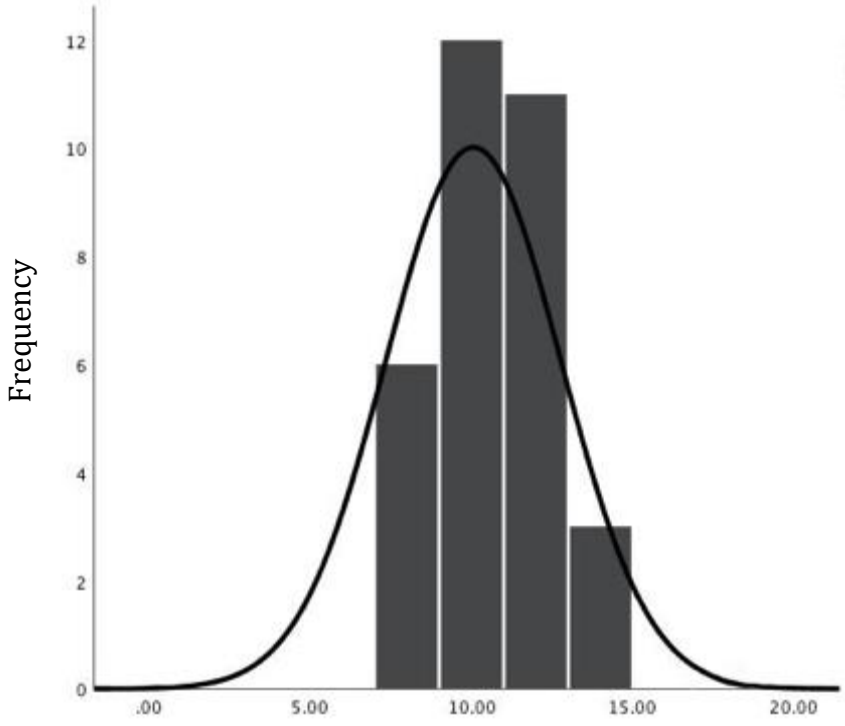
Appendix 3.1 First pilot results of biological knowledge of genetics.



Appendix 3.2 Second pilot study of biological knowledge of genetics.



Appendix 3.3 Pilot results of clinical knowledge of genetics.



Appendix 3.4 Breakdown of the recruitment method.

Response	Organisation/Person	Type of contact	Means of distribution
Distributed (*n* = 23)	Dr Anna Middleton	Academic	Via Twitter
	Union UCL BME Students' Network	Student group	Distributed via Facebook
	SU BME Network	Student group	Distributed via Facebook
	Teacher at primary school	Personal	Distributed via email around school
	University of Bristol	Academic	Distributed via Twitter
	Bristol Alumni	Academic	Distributed via LinkedIn Alumni page
	The Academic Midwife	Academic	Distributed via Facebook
	VCS Cymru	Volunteering group	Distributed via email
	Redland & Cotham Amenities Society	Community/Volunteering group	Distributed via email
	disAbility Cornwall & Isles of Scilly	Community/Volunteering group	Distributed via email
	The Dracaena Centre	Community/Volunteering group	Distributed via email
	Crown Prosecution Service Section Council	Personal	Distributed via email
	Friends of High Town chair	Community/Volunteering group	Distributed via email and personal twitter
	Science Museum, London	Science organisation	Distributed via social media
	Headteacher at secondary school	Personal	Distributed via email around school
	Bristol Astronomical Society	Community/Volunteering Group	Distributed via email

	Bristol Shiplovers Society	Community/Volunteering group	Distributed via email
	NoFitState Cardiff	Circus company	Distributed via email
	Stephen's Scown law firm	Personal	Distributed via email
	CoppaFeel!	Personal	Distributed via email
	OVAID	Charity	Distributed via email
	Children Change Colombia	Charity	Distributed via email
	The Winnie Mabaso Foundation	Charity	Distributed via email
Declined (*n* = 11)	We The Curious	Science organisation	N/A
	National Osteoporosis Society	Charity	N/A
	Parkinson's UK	Charity	N/A
	Devon Wildlife Trust	Charity	N/A
	Cornwall Wildlife Trust	Charity	N/A
	Bath Heritage Watchdog	Community/Volunteering group	N/A
	MS Society	Charity	N/A
	Avon Organic Group	Community/Volunteering group	N/A
	Refugee Action	Charity	N/A
	Bristol Industrial Archaeological Society	Community/Volunteeringgroup	N/A
	Friends of the Downs and Avon Gorge	Community/Volunteering group	N/A
No response (*n* = 356)	Transition Bath	Community/Volunteering group	N/A
	The New Somerset and Dorset Railway	Community/Volunteering group	N/A
	Bath Area Play Project	Community/Volunteering group	N/A
	Julian House	Community/Volunteering group	N/A
	NRAS / Bath & West Wilts branch	Community/Volunteering group	N/A
	National Rheumatoid Arthritis Society	Community/Volunteering group	N/A
	RICE / Memory Clinics	Community/Volunteering group	N/A

	St John's Ambulance	Community/Volunteering group	N/A
	The Carers' Centre (Bath and North East Somerset)	Community/Volunteering group	N/A
	The Integrative Health Trust	Community/Volunteering group	N/A
	Arthritis Care – Bath Support Group	Community/Volunteering group	N/A
	Doing Good Leeds	Community/Volunteering group	N/A
	Blx Better Leeds Communities	Community/Volunteering group	N/A
	Leeds Mencap	Community/Volunteering group	N/A
	Improving Lives Plymouth	Community/Volunteering group	N/A
	Plymouth Environmental Action	Community/Volunteering group	N/A
	Age UK Plymouth	Community/Volunteering group	N/A
	The zone Plymouth	Community/Volunteering group	N/A
	Keyham Methodist Community Centre	Community/Volunteering group	N/A
	NCVO Volunteering	Community/Volunteering group	N/A
	FoodCycle	Community/Volunteering group	N/A
	Buckinghamshire County Council	Community/Volunteering group	N/A
	Cambridgeshire County Council	Community/Volunteering group	N/A
	Dorset City Council	Community/Volunteering group	N/A
	East Sussex County Council	Community/Volunteering group	N/A
	Bournemouth Borough Council	Community/Volunteering group	N/A
	Age Concern – Bournemouth	Community/Volunteering group	N/A
	Brendon Care	Community/Volunteering group	N/A
	Books on Wheels	Community/Volunteering group	N/A
	Bournemouth Council for Voluntary Service	Community/Volunteering group	N/A

	British Red Cross	Community/Volunteering group	N/A
	Christchurch Volunteer Bureau	Community/Volunteering group	N/A
	Compass & Focus Office	Community/Volunteering group	N/A
	Contact the Elderly	Community/Volunteering group	N/A
	Dorset Blind Association	Community/Volunteering group	N/A
	Dorset Community Action – Head Office	Community/Volunteering group	N/A
	Stroke Association	Community/Volunteering group	N/A
	Streetwise Safety Centre	Community/Volunteering group	N/A
	Elf Exeter Leukaemia	Community/Volunteering group	N/A
	Exeter City Farm	Community/Volunteering group	N/A
	Devon Wildlife Trust	Community/Volunteering group	N/A
	Exeter Student Volunteers	Community/Volunteering group	N/A
	Devon County Council	Community/Volunteering group	N/A
	Volunteer Cornwall	Community/Volunteering group	N/A
	Cornwall Rural Community Charity	Community/Volunteering group	N/A
	Cornwall Social Group	Community/Volunteering group	N/A
	Open Gardens Volunteers	Community/Volunteering group	N/A
	Team Green!	Community/Volunteering group	N/A
	Wildlife Watch Volunteer Opportunities	Community/Volunteering group	N/A
	Armed forces and Veterans Breakfast	Community/Volunteering group	N/A
	A Band Of Brothers	Community/Volunteering group	N/A
	Bodmin Day Centre	Community/Volunteering group	N/A
	British Red Cross – Connecting Communities Project	Community/Volunteering group	N/A

	C Fylm	Community/Volunteering group	N/A
	CAMEO – Saltash Live at Home	Community/Volunteering group	N/A
	Centre Of Pendeen	Community/Volunteering group	N/A
	Contact the Elderly	Community/Volunteering group	N/A
	Cornwall Neighbourhoods for Change	Community/Volunteering group	N/A
	Cornwall Tech Jam – Brithday Bash	Community/Volunteering group	N/A
	Creative writing workshop	Community/Volunteering group	N/A
	Echo Centre, Day Resource Centre	Community/Volunteering group	N/A
	Hayle Day Care Centre	Community/Volunteering group	N/A
	Lostwithiel Messy Church	Community/Volunteering group	N/A
	Mens Pub Lunch Group – Saltash Live At Home	Community/Volunteering group	N/A
	The WI: Inspiring Women	Community/Volunteering group	N/A
	Park View	Community/Volunteering group	N/A
	Young farmers club	Community/Volunteering group	N/A
	Treverbyn community hall	Community/Volunteering group	N/A
	Royal Voluntary Service – social and lunch club	Community/Volunteering group	N/A
	Southampton Voluntary Services	Community/Volunteering group	N/A
	Pavillion on the Park	Community/Volunteering group	N/A
	Velmore Community Centre	Community/Volunteering group	N/A
	Community First	Community/Volunteering group	N/A
	UNIT 12	Community/Volunteering group	N/A
	Wallington Village Community Association	Community/Volunteering group	N/A

	Carroll Centre	Community/Volunteering group	N/A
	Somers Town Community Association	Community/Volunteering group	N/A
	John Pounds Centre	Community/Volunteering group	N/A
	Landport Community Centre	Community/Volunteering group	N/A
	Buckland Community Centre	Community/Volunteering group	N/A
	Fratton Community Centre	Community/Volunteering group	N/A
	The Guinness partnership	Community/Volunteering group	N/A
	Havelock Community Centre	Community/Volunteering group	N/A
	Hayling Island	Community/Volunteering group	N/A
	Get Involved Slough	Community/Volunteering group	N/A
	Edible High Town	Community/Volunteering group	N/A
	Hightown Honeys (WI)	Community/Volunteering group	N/A
	Norwood Community Group	Community/Volunteering group	N/A
	St Joseph's Hospice	Community/Volunteering group	N/A
	Waterloo Community Group	Community/Volunteering group	N/A
	The Sock Mob	Community/Volunteering group	N/A
	London and West Middlesex National Trust	Community/Volunteering group	N/A
	Hands on London	Community/Volunteering group	N/A
	City of London	Community/Volunteering group	N/A
	Somers Town Community Association	Community/Volunteering group	N/A
	Maiden Lane Community Centre	Community/Volunteering group	N/A
	St Mary's (Eltham) Community Complex	Community/Volunteering group	N/A

	Coram's Field	Community/Volunteering group	N/A
	Self Management UK	Community/Volunteering group	N/A
	Alford House	Community/Volunteering group	N/A
	Vauxhall City Farm	Community/Volunteering group	N/A
	Kings corner project	Community/Volunteering group	N/A
	EMCA	Community/Volunteering group	N/A
	The Feathers Association	Community/Volunteering group	N/A
	Sky Way London	Community/Volunteering group	N/A
	Eastside	Community/Volunteering group	N/A
	Inspire London college	Community/Volunteering group	N/A
	Oaktree Community Centre	Community/Volunteering group	N/A
	Highbury Roundhouse	Community/Volunteering group	N/A
	Streatham Youth and Community Trust	Community/Volunteering group	N/A
	Elizabeth House	Community/Volunteering group	N/A
	Kingsley Hall Community Centre	Community/Volunteering group	N/A
	New Public Services Isledon	Community/Volunteering group	N/A
	Caius House Youth	Community/Volunteering group	N/A
	Hargrave Hall Community Association	Community/Volunteering group	N/A
	TowerProject	Community/Volunteering group	N/A
	Copleston Centre	Community/Volunteering group	N/A
	The Space	Community/Volunteering group	N/A
	Holly Lodge Community Centre	Community/Volunteering group	N/A
	Choices	Community/Volunteering group	N/A

	Kingsgate Community Centre	Community/Volunteering group	N/A
	HBC Community Centre	Community/Volunteering group	N/A
	Cardinal Heenan Centre	Community/Volunteering group	N/A
	Muslim Welfare House	Community/Volunteering group	N/A
	Hampstead Community Centre	Community/Volunteering group	N/A
	Islington Arts Factory	Community/Volunteering group	N/A
	Norwood Community Group	Community/Volunteering group	N/A
	St Mark's Community Centre	Community/Volunteering group	N/A
	Poplar Harca 20	Community/Volunteering group	N/A
	WFC Working for the Community	Community/Volunteering group	N/A
	Lewisham Indochinese Community Centre	Community/Volunteering group	N/A
	Kelston Club & Study Centre	Community/Volunteering group	N/A
	Alpha Grove	Community/Volunteering group	N/A
	Peter House Centre	Community/Volunteering group	N/A
	Brand New Start	Community/Volunteering group	N/A
	St Faith's Centre	Community/Volunteering group	N/A
	Postive Network	Community/Volunteering group	N/A
	AccessAble	Community/Volunteering group	N/A
	Global Woman.co	Community/Volunteering group	N/A
	St Mary's Tottenham	Community/Volunteering group	N/A
	Aston Mansfield	Community/Volunteering group	N/A
	Street Vibes	Community/Volunteering group	N/A
	LondonAssembly	Community/Volunteering group	N/A

	Brand New Start	Community/Volunteering group	N/A
	St Faith's Centre	Community/Volunteering group	N/A
	POstive Network	Community/Volunteering group	N/A
	AccessAble	Community/Volunteering group	N/A
	Global Woman.co	Community/Volunteering group	N/A
	St Mary's Tottenham	Community/Volunteering group	N/A
	Aston Mansfield	Community/Volunteering group	N/A
	Street Vibes	Community/Volunteering group	N/A
	LondonAssembly	Community/Volunteering group	N/A
	Oxfordshire Community & Voluntary Action	Community/Volunteering group	N/A
	Arts Society Oxford	Community/Volunteering group	N/A
	The Gatehouse	Community/Volunteering group	N/A
	The Community Action Groups	Community/Volunteering group	N/A
	Church Mission Society	Community/Volunteering group	N/A
	Oxfordshire Young Farmers	Community/Volunteering group	N/A
	Leys Community Development Initiative	Community/Volunteering group	N/A
	Oxford Union	Community/Volunteering group	N/A
	Watford Community Housing	Community/Volunteering group	N/A
	Mind Hertfordshire Network	Community/Volunteering group	N/A
	Teamherts	Community/Volunteering group	N/A
	Three Rivers District Council	Community/Volunteering group	N/A
	Watford Mencap	Community/Volunteering group	N/A
	The Vineyard	Community/Volunteering group	N/A

	Cedars youth and community centre	Community/Volunteering group	N/A
	Expressions Academy	Community/Volunteering group	N/A
	Harrow Monitoring Group	Community/Volunteering group	N/A
	iGNITE!	Community/Volunteering group	N/A
	The Pavilion	Community/Volunteering group	N/A
	Wembley PIWC	Community/Volunteering group	N/A
	Elm Court Youth and Community Centre	Community/Volunteering group	N/A
	WEC Youth Camps	Community/Volunteering group	N/A
	Kingsgate Community Centre	Community/Volunteering group	N/A
	Hampstead Community Centre	Community/Volunteering group	N/A
	Willen Hospice	Community/Volunteering group	N/A
	Age UK Milton Keynes	Community/Volunteering group	N/A
	Inter-Action UK	Community/Volunteering group	N/A
	Conniburrow Community Association	Community/Volunteering group	N/A
	Willen Hospice	Community/Volunteering group	N/A
	Liverpool City Council	Community/Volunteering group	N/A
	LCVS	Community/Volunteering group	N/A
	Birkenhead Youth Club	Community/Volunteering group	N/A
	Open Door Centre	Community/Volunteering group	N/A
	The Cross Birkenhead	Community/Volunteering group	N/A
	Bootle Christ Church Youth & Community Centre	Community/Volunteering group	N/A
	Merseyside Youth Association	Community/Volunteering group	N/A
	Brunswick Youth and Community Centre	Community/Volunteering group	N/A

	St Mary's Millennium Centre	Community/Volunteering group	N/A
	Centre 63	Community/Volunteering group	N/A
	Thornton Hough Village Hall	Community/Volunteering group	N/A
	Halton Borough Council	Community/Volunteering group	N/A
	Mencap Liverpool	Community/Volunteering group	N/A
	KCVS	Community/Volunteering group	N/A
	Netherton FeelGood Factory	Community/Volunteering group	N/A
	Info Buzz	Community/Volunteering group	N/A
	Vision 21	Community/Volunteering group	N/A
	Lions Club international	Community/Volunteering group	N/A
	Marah	Community/Volunteering group	N/A
	Sheppard House	Community/Volunteering group	N/A
	Neighbourhood Watch Conduit-Falkner	Community/Volunteering group	N/A
	Gloucestershire Federation of Young Farmers' Clubs	Community/Volunteering group	N/A
	Gloucester Youth Support Team	Community/Volunteering group	N/A
	Shurdington	Community/Volunteering group	N/A
	Gloucestershire County Council	Community/Volunteering group	N/A
	The Door Unlocking Potential	Community/Volunteering group	N/A
	Heart of Priors Park	Community/Volunteering group	N/A
	Love Woodmancote	Community/Volunteering group	N/A
	Ipswich Pentecostal Church	Community/Volunteering group	N/A
	Suffol Young Farmers	Community/Volunteering group	N/A
	Harwich Horse Rangers Association	Community/Volunteering group	N/A

	Pathways Care Group	Community/Volunteering group	N/A
	Anglia Care Trust	Community/Volunteering group	N/A
	Northamptonshire Dyslexia Association	Community/Volunteering group	N/A
	Northampton Sikhs	Community/Volunteering group	N/A
	Abington Community Centre	Community/Volunteering group	N/A
	Duston Parish Council	Community/Volunteering group	N/A
	Umbrella Fair Organisation	Community/Volunteering group	N/A
	The Pastures Community Centre	Community/Volunteering group	N/A
	Parklands Community Centre	Community/Volunteering group	N/A
	Grane Park Community centre	Community/Volunteering group	N/A
	Storehouse church	Community/Volunteering group	N/A
	Hardingstone Village Hall	Community/Volunteering group	N/A
	The Centre at Mawsley	Community/Volunteering group	N/A
	Glamis Hall for All	Community/Volunteering group	N/A
	Victoria Centre	Community/Volunteering group	N/A
	Daventry Community Centre	Community/Volunteering group	N/A
	WACA	Community/Volunteering group	N/A
	Olney Town council	Community/Volunteering group	N/A
	New Start 4 u	Community/Volunteering group	N/A
	Koco Community Resource Centre	Community/Volunteering group	N/A
	Wild Earth	Community/Volunteering group	N/A
	Jubilee Crescent Community Centre	Community/Volunteering group	N/A
	Canley Community centre	Community/Volunteering group	N/A
	Daimler Green Community Centre	Community/Volunteering group	N/A

	Bedworth Heath Community Centre	Community/Volunteering group	N/A
	St Francis Community Centre	Community/Volunteering group	N/A
	Arley and St.Michael's Community Centre	Community/Volunteering group	N/A
	Stockingford Community Centre	Community/Volunteering group	N/A
	Warwickgates Community Centre	Community/Volunteering group	N/A
	Westfield Community Centre	Community/Volunteering group	N/A
	Chase Meadow Community Centre	Community/Volunteering group	N/A
	Sydenham Neighbourhood Initiatives	Community/Volunteering group	N/A
	The gap	Community/Volunteering group	N/A
	Benn Partnership Centre	Community/Volunteering group	N/A
	BVC community & conference centre	Community/Volunteering group	N/A
	Graham Adams Centre	Community/Volunteering group	N/A
	Rugby West Indian Association social club & community centre (fb msg)	Community/Volunteering group	N/A
	Bentley Heath Community centre	Community/Volunteering group	N/A
	BHLC	Community/Volunteering group	N/A
	Hinckley Natural History Museum	Community/Volunteering group	N/A
	Mancetter Memorial Hall	Community/Volunteering group	N/A
	Churches of Arden	Community/Volunteering group	N/A
	Cyrenians	Community/Volunteering group	N/A
	St Basils	Community/Volunteering group	N/A
	The Salvation Army	Community/Volunteering group	N/A
	East Birmingham Community Forum	Community/Volunteering group	N/A

	The Midlands Greek and Cypriot Association	Community/Volunteering group	N/A
	St. Wilfrid's Community Centre	Community/Volunteering group	N/A
	Warley Woods Community Trust	Community/Volunteering group	N/A
	Shenley Court Hall	Community/Volunteering group	N/A
	The Brandwood Centre	Community/Volunteering group	N/A
	Quinborne Community Centre	Community/Volunteering group	N/A
	Rowheath Pavilion	Community/Volunteering group	N/A
	Endo ball	Community/Volunteering group	N/A
	Welcome Change	Community/Volunteering group	N/A
	Tekio Gemu	Community/Volunteering group	N/A
	HPP	Community/Volunteering group	N/A
	Clubmark Old Hill CC	Community/Volunteering group	N/A
	Midland Youth Jazz Orchestra	Community/Volunteering group	N/A
	Brierley Hill Project	Community/Volunteering group	N/A
	The Parish Centre at St Chad's	Community/Volunteering group	N/A
	BVSC	Community/Volunteering group	N/A
	Additional Curates Society	Community/Volunteering group	N/A
	Bangladesh Women's Empowerment Resource Centre	Community/Volunteering group	N/A
	The Kaleidoscope Plus Group	Community/Volunteering group	N/A
	Oscar Sandwell Co. Ltd	Community/Volunteering group	N/A
	Confederation of Bangladeshi Organisations	Community/Volunteering group	N/A
	The Bircham Centre	Community/Volunteering group	N/A

	Belvedere Community Centre	Community/Volunteering group	N/A
	Harford Community Centre	Community/Volunteering group	N/A
	Cringleford Parish Council	Community/Volunteering group	N/A
	Clover Hill Village Farm	Community/Volunteering group	N/A
	Lingwood Village Hall	Community/Volunteering group	N/A
	Jubilee Family Centre	Community/Volunteering group	N/A
	Hickling Barn	Community/Volunteering group	N/A
	Reedham Village Hall	Community/Volunteering group	N/A
	Peterborough City Council	Community/Volunteering group	N/A
	Italian Community Association	Community/Volunteering group	N/A
	Gauntlet Auto Project LTD	Community/Volunteering group	N/A
	The Mandela Community Centre	Community/Volunteering group	N/A
	Mickleover Community Centre	Community/Volunteering group	N/A
	Stapleford Community Group	Community/Volunteering group	N/A
	Nuthall Parish Council	Community/Volunteering group	N/A
	Hidden Disabilities	Community/Volunteering group	N/A
	Umbrella - Embracing Abilities	Community/Volunteering group	N/A
	Eastwood Volunteer Bureau	Community/Volunteering group	N/A
	St Albans community centre	Community/Volunteering group	N/A
	Bradwell Parish Council	Community/Volunteering group	N/A
	The Victoria Centre	Community/Volunteering group	N/A
	Changes	Community/Volunteering group	N/A
	Royal Voluntary Services	Community/Volunteering group	N/A
	The Donna Louise	Community/Volunteering group	N/A

	Oak tree farm rural project	Community/Volunteering group	N/A
	Voluntary action sheffield	Community/Volunteering group	N/A
	Emmaus Sheffield	Community/Volunteering group	N/A
	Tassibee	Community/Volunteering group	N/A
	Ashgate Hospicecare	Community/Volunteering group	N/A
	St Paul's church	Community/Volunteering group	N/A
	Richmond Church	Community/Volunteering group	N/A
	The Link Community	Community/Volunteering group	N/A
	Woodhouse and District Community Forum	Community/Volunteering group	N/A
	Manchester Carers Forum	Community/Volunteering group	N/A
	Cornerstone	Community/Volunteering group	N/A
	Salford CVS	Community/Volunteering group	N/A
	ROC	Community/Volunteering group	N/A
	Just add CIC	Community/Volunteering group	N/A
	Manchester Youth Zone	Community/Volunteering group	N/A
	Rising Stars	Community/Volunteering group	N/A
	Oak Community Development	Community/Volunteering group	N/A
	Bury stars	Community/Volunteering group	N/A
	The Mosses Community	Community/Volunteering group	N/A
	HeartLift	Community/Volunteering group	N/A
	The Hub	Community/Volunteering group	N/A
	Castlemere	Community/Volunteering group	N/A
	Re'new	Community/Volunteering group	N/A

	Barca-Leeds	Community/Volunteering group	N/A
	YMCA Leeds	Community/Volunteering group	N/A
	LS14 Trust	Community/Volunteering group	N/A
	The Youth Association	Community/Volunteering group	N/A
	Prism Youth Project	Community/Volunteering group	N/A
	Manningham Mills Sports & Community Association,	Community/Volunteering group	N/A
	Cottingley Community Centre	Community/Volunteering group	N/A
	Otley Courthouse	Community/Volunteering group	N/A
	York Against Cancer	Community/Volunteering group	N/A
	York Centre Voluntary Service	Community/Volunteering group	N/A
	Burnley Pendle & Rossendale Council for Voluntary Service	Community/Volunteering group	N/A
	Bristol Orbit Club	Community/Volunteering group	N/A
	Disabled Motoring UK	Community/Volunteering group	N/A
	WECIL	Community/Volunteering group	N/A
	British Cactus & Succulent Society: Bristol Branch	Community/Volunteering group	N/A
	Bristol Food Network	Community/Volunteering group	N/A
	Park Work	Community/Volunteering group	N/A
	Royal Horticulture Society	Community/Volunteering group	N/A
	Goodgym	Community/Volunteering group	N/A
	Urban Buzz Bristol	Community/Volunteering group	N/A
	Bristol Zero Tolerance	Community/Volunteering group	N/A
	Southville Gardening Club	Community/Volunteering group	N/A

	Golden Hill Community	Community/Volunteering group	N/A
	Walled Kitchen Gardens Network	Community/Volunteering group	N/A
	Arnos Vale	Community/Volunteering group	N/A
	Friends of Horfield Common	Community/Volunteering group	N/A
	Friends of Old Sneed Park Nature Reserve	Community/Volunteering group	N/A
	Bristol and Bath intervarsity Club	Community/Volunteering group	N/A
	Bristol Social group	Community/Volunteering group	N/A
	Bristol Dog training society	Community/Volunteering group	N/A
	Bristol supper club	Community/Volunteering group	N/A
	Bristol & South Gloucestershire Stationary Engine	Community/Volunteering group	N/A
	National Vintage Tractor & Engine Club	Community/Volunteering group	N/A
	Bristol Skeptics Society	Community/Volunteering group	N/A
	Yogawest	Community/Volunteering group	N/A
	Bristol & Bath Lug	Community/Volunteering group	N/A
	Association for Roman Archaeology	Community/Volunteering group	N/A
	Bristol Industrial Archaeological Society	Community/Volunteering group	N/A
	Friends of BMGA, Supporting Bristol Museums, Galleries & Archives	Community/Volunteering group	N/A
	National Association of Re-enactment Societies	Community/Volunteering group	N/A
	Bristol Aero Club	Community/Volunteering group	N/A
	Bristol & Wessex Aeroplane Club	Community/Volunteering group	N/A
	Bristol Hot Air Balloon Society	Community/Volunteering group	N/A
	Bristol Concert Wind Band	Community/Volunteering group	N/A

	Bristol Hippodrome Choir	Community/Volunteering group	N/A
	West Bristol Orchestra	Community/Volunteering group	N/A
	Redland Wind Band	Community/Volunteering group	N/A
	City of Bristol College	Community/Volunteering group	N/A
	South Gloucestershire and Stroud College	Community/Volunteering group	N/A
	City of Bristol College	Community/Volunteering group	N/A
	CUSU BME Campaign	Student group	N/A
	Bristol University BME Success	Student group	N/A
	The BME Collective	Campaign	N/A

Appendix 3.5 Questions eliminated from each section following Cronbach's alpha test of reliability.

[Section 1 - Biological knowledge of genetics]

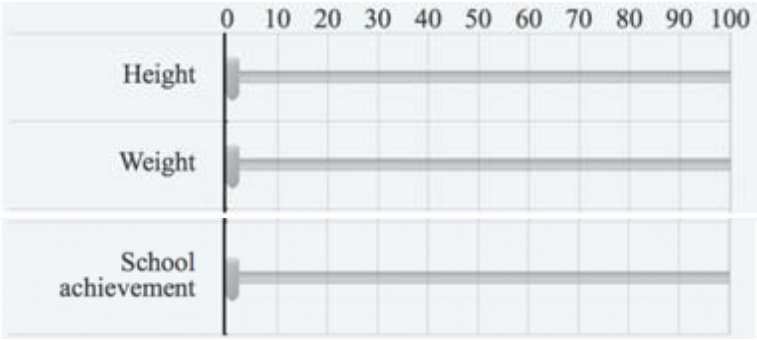
13. If you share 50% of your DNA variants with your sister, how much do you share with your cousin?

- 50%
- 25%
- 12.5%
- 8%

16. Approximately how many genes does the human DNA code contain?

- 2,000
- 1 million
- 3 billion
- 20,000

19, 20, 22. On a scale of 0-100 how important do you think genetic differences are between people in explaining individual differences in the following traits (with 100 = only genetic differences can explain individual differences in traits):



[Clinical knowledge of genetics in healthcare]

2 There are many common diseases where the study of genetics can show the road to better treatment

- True
- False

3. The use of genetics in healthcare can mean that expensive drugs are only given to those who will benefit from them

- True

- False

4. It is against the law for a doctor to **not** disclose a patient's genetic test results to their close relatives as, given their genetic relatedness, the results may also concern them
- True
- False

5. Currently, treatment for cancer can include a genetic diagnosis to show if a tumour might respond to a certain treatment
- True
- False

6. The use of patients' genetic data to deliver targeted therapies is already changing people's lives
- True
- False

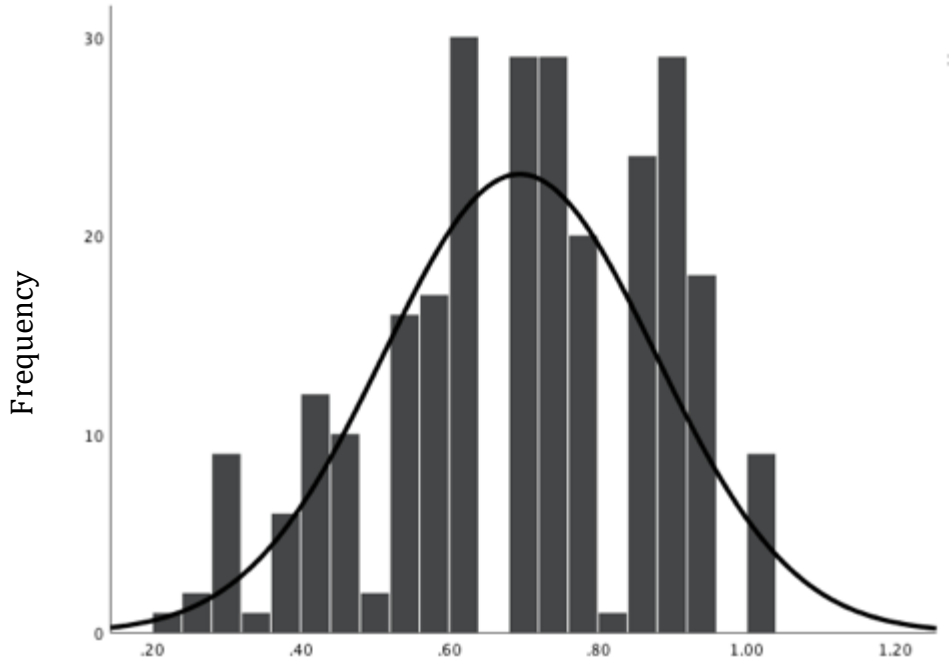
7. A patient will only speak with a genetic counsellor if their genetic test result indicates they have a genetic predisposition to a disease
- True
- False

8. Through a number of genetic testing techniques, it is possible to detect genetic abnormalities in an unborn child
- True
- False

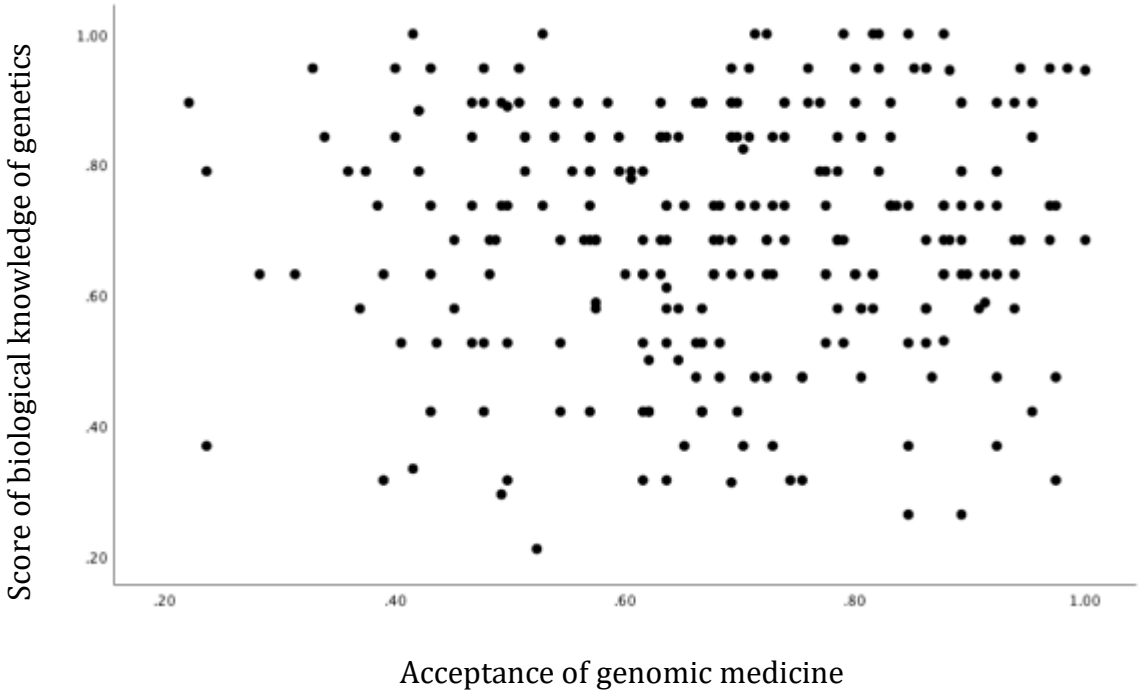
11. At present in the UK, new born infants are tested for certain genetic traits
- True
- False

Appendix 3.6 Results for the data screening process.

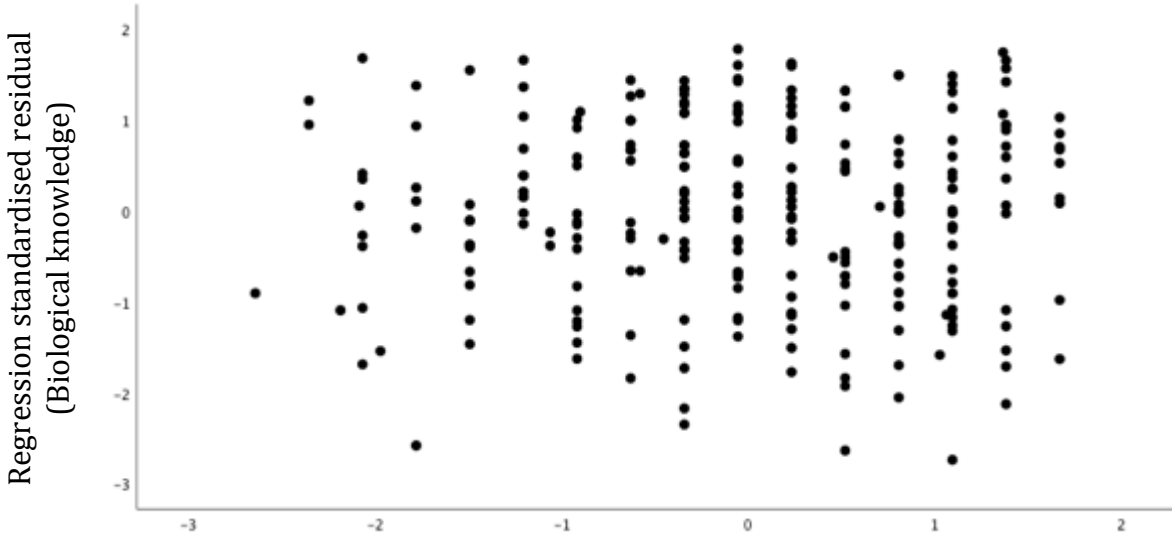
Normality plot of the biological knowledge of genetics measure.



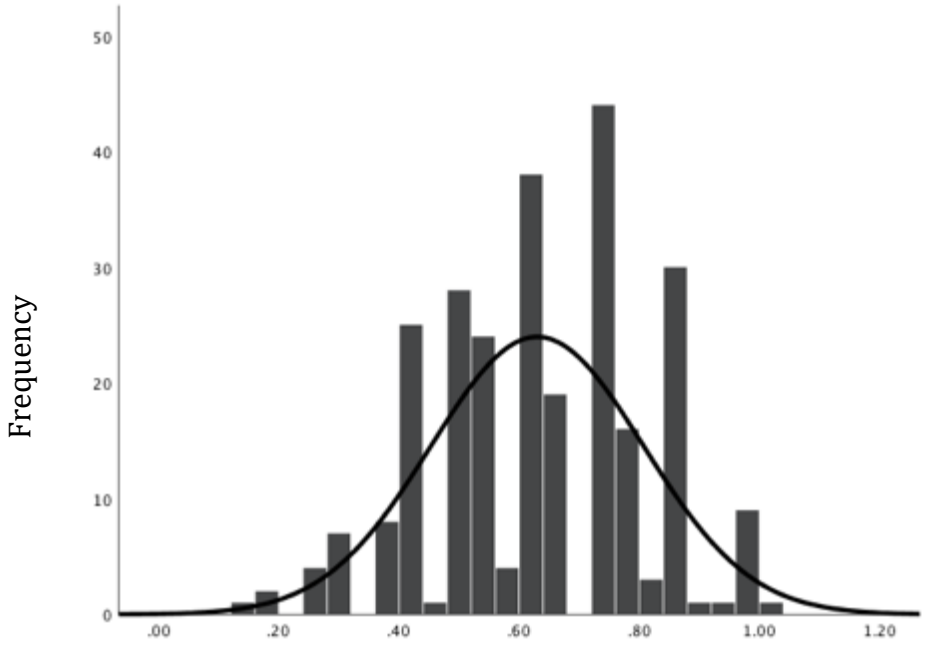
Linearity plot between the biological knowledge of genetics measure and acceptance of genomic medicine measure.



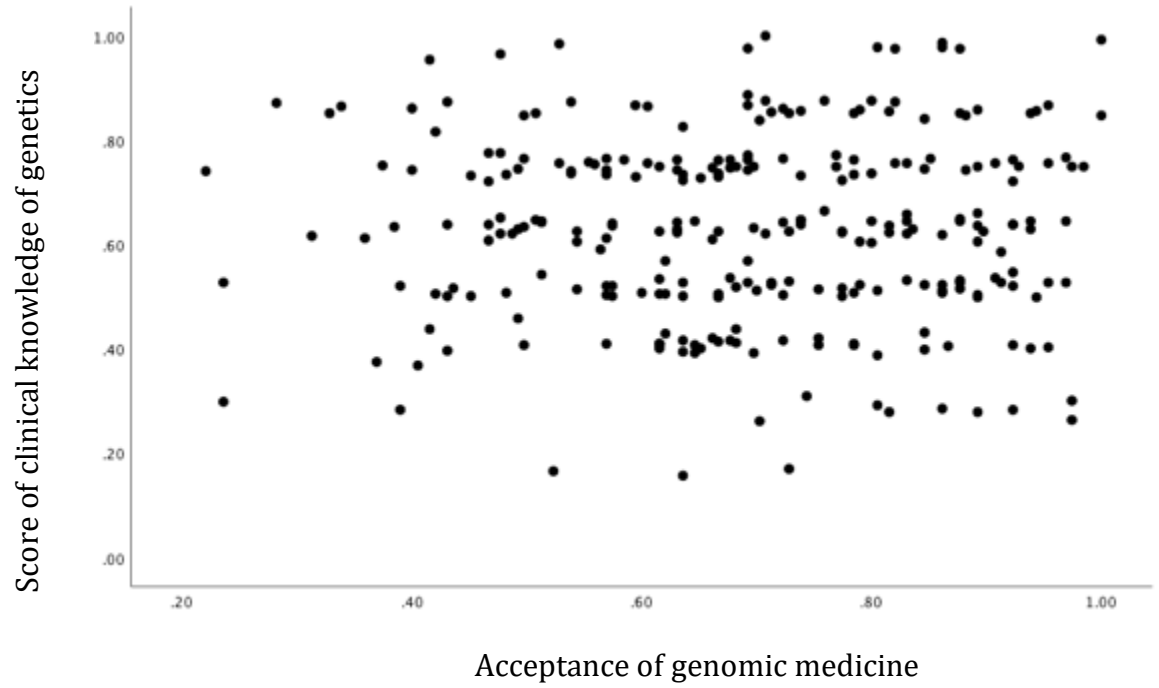
Homoscedasticity plot of the biological knowledge of genetics measure.



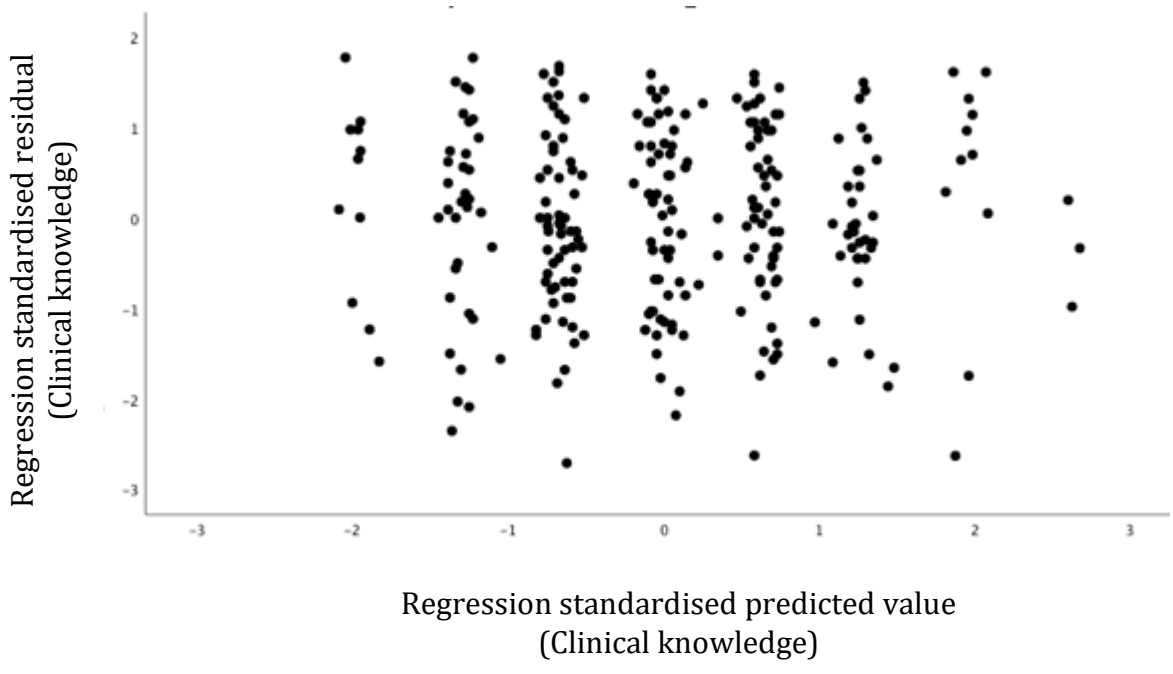
Normality plot of the clinical knowledge of genetics measure.



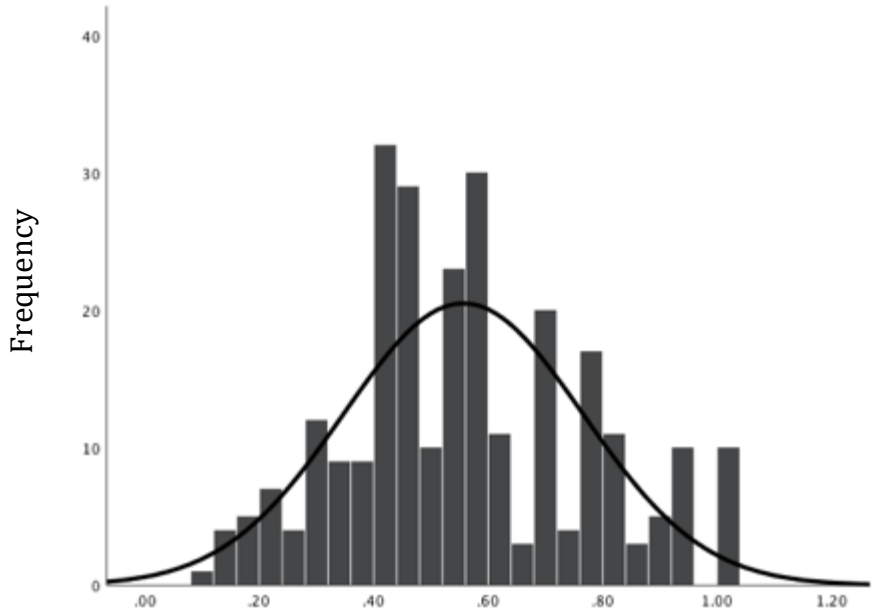
Linearity plot of the clinical knowledge of genetics measure.



Homoscedasticity plot of the clinical knowledge of genetics measure.

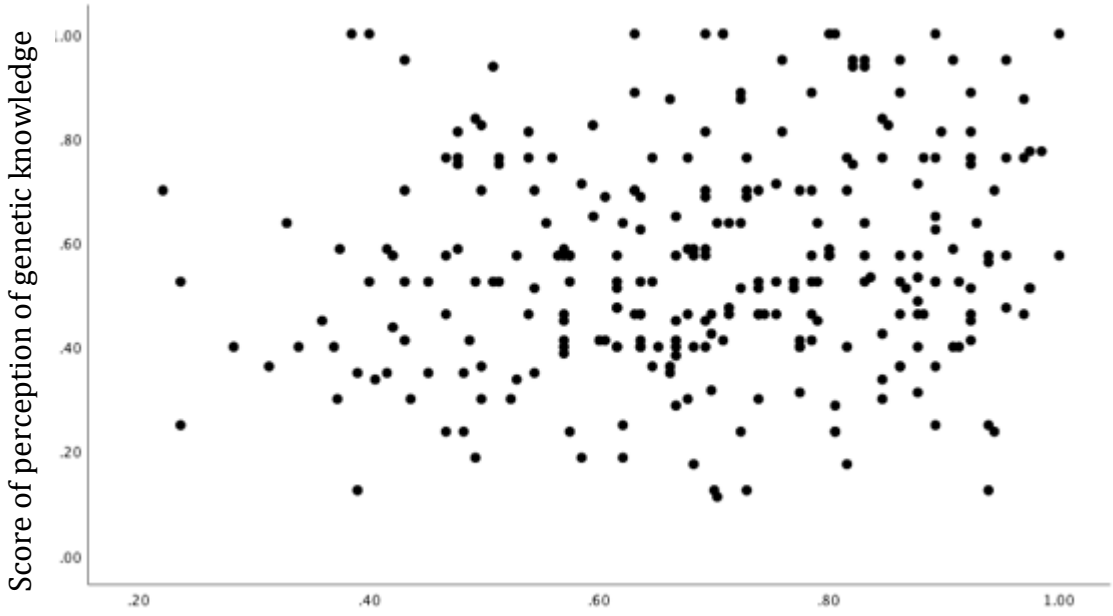


Normality plot of the perception of genetic knowledge measure.



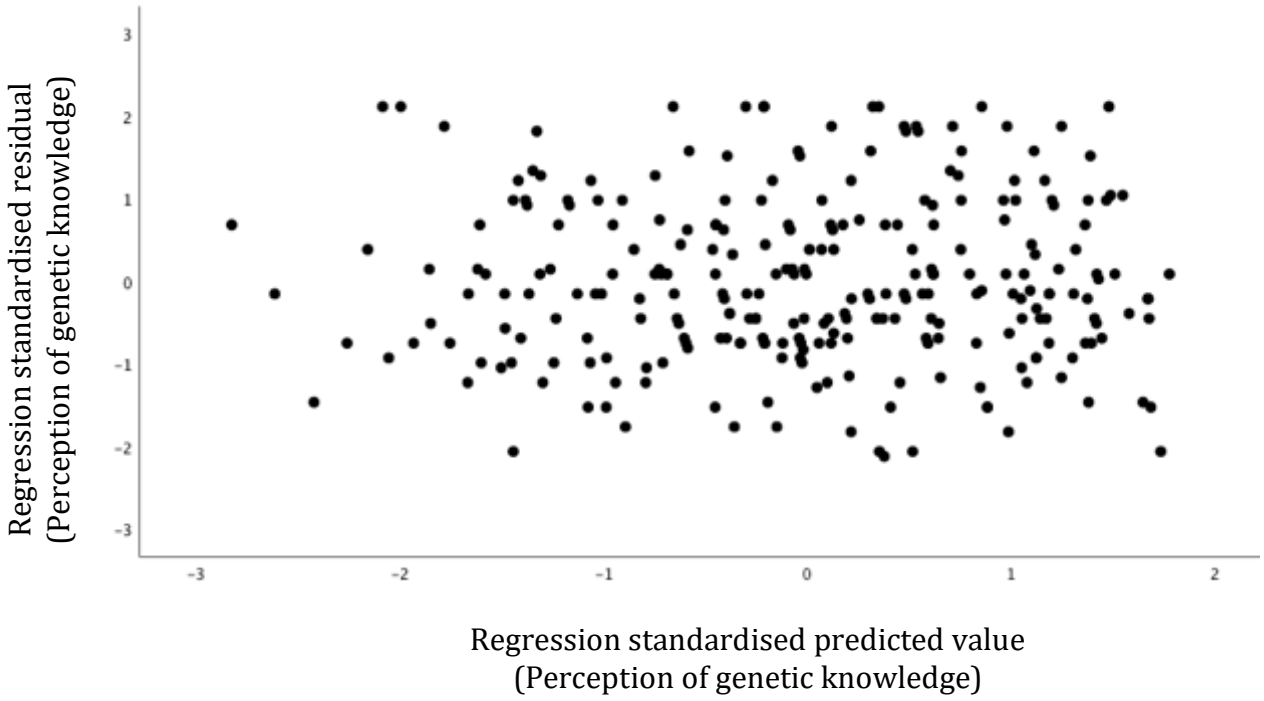
Score of perception of genetic knowledge

Linearity plot of the perception of genetic knowledge measure.

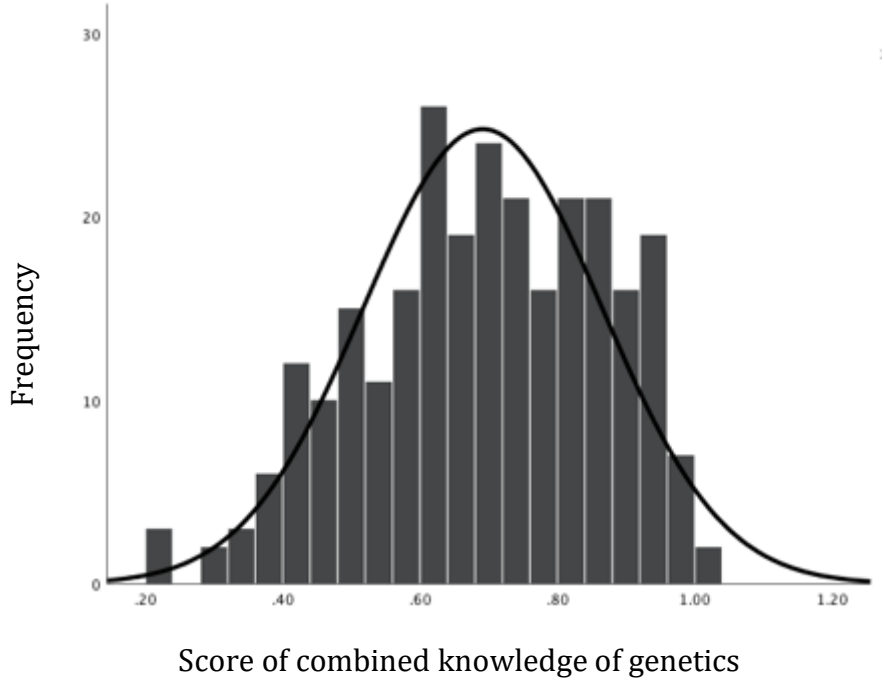


Acceptance of genomic medicine

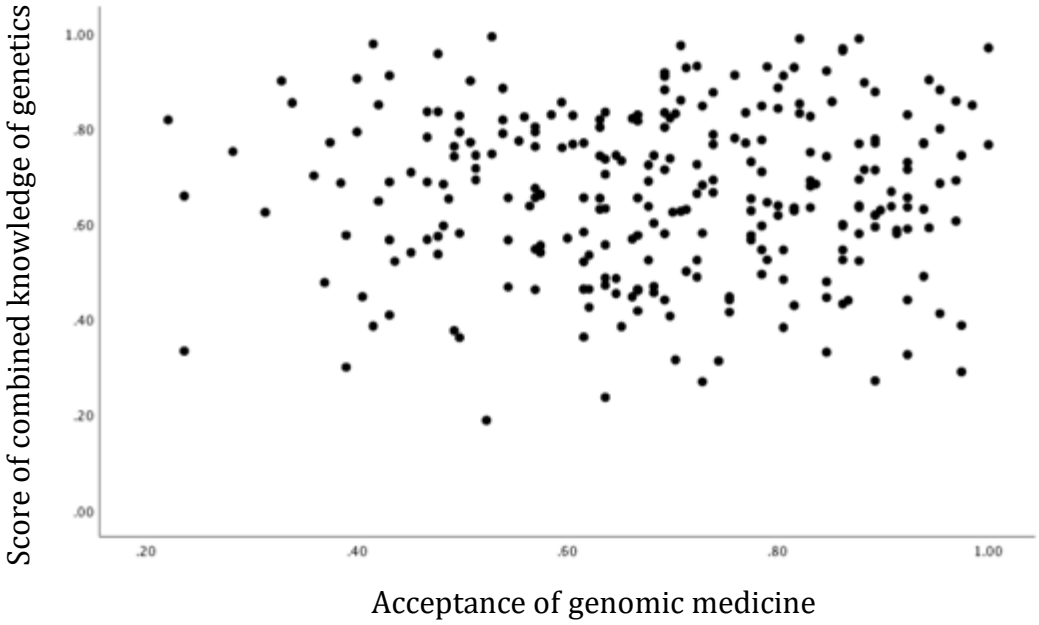
Homoscedasticity plot of the perception of genetic knowledge measure.



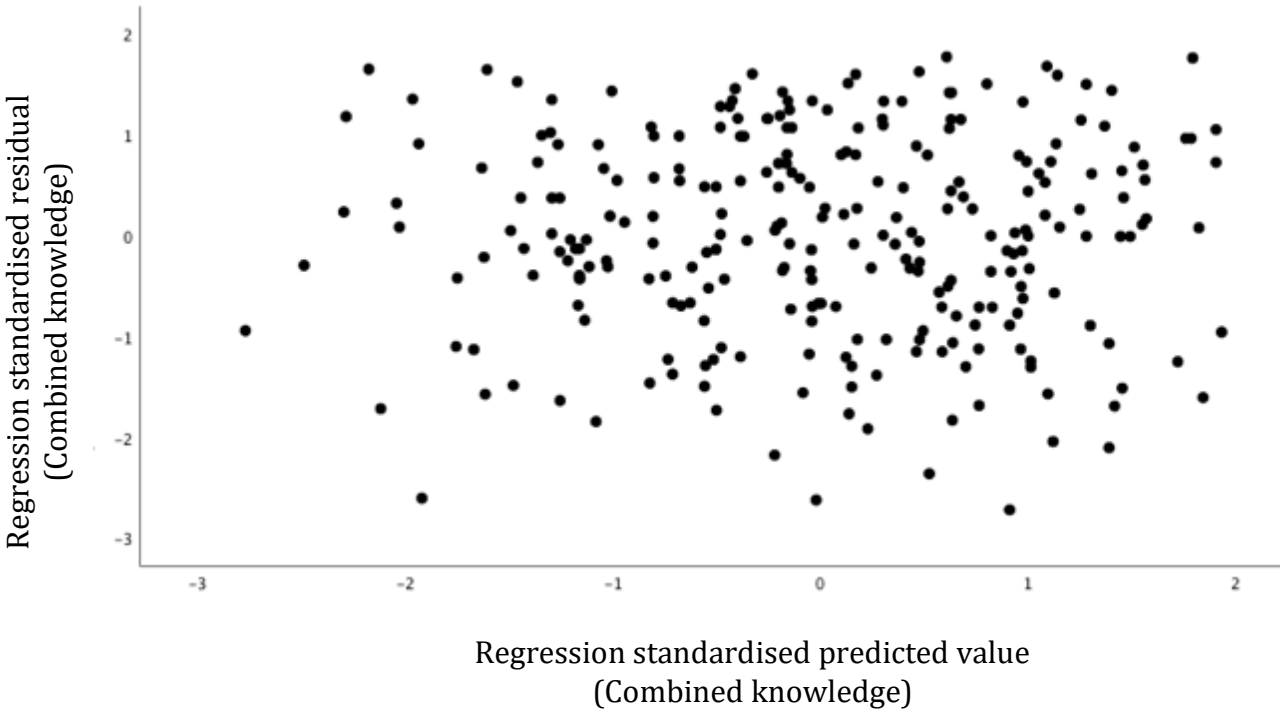
Normality plot of the combined knowledge measure.



Linearity plot of the combined knowledge measure.



Homoscedasticity plot of the combined knowledge measure.



Appendix 4 Application, conditional letter, our response, and favourable opinion letter.

Faculty of Science Human Research Ethics Committee
APPLICATION FOR RESEARCH ETHICS APPROVAL

University of BRISTOL

1. **Title of the research:**
Thoughts from the UK public on the use of genetics in healthcare: Concerns, expectations, and the provision of decision-relevant information

2. **Name of Applicant, with their job title:**
Helena Davies, Postgraduate research student

3. **Name of Supervisor (if applicant is a postgraduate or undergraduate student), with their job title:**
Dr Claire Haworth, Reader in Behavioural Genetics

4. **Other investigator(s) involved, with their job title:**
Dr Robyn Wootton, Postdoctoral researcher
Dr Oliver Davis, Reader in Statistical Genetics

5. **Source of funding and grant code:**
I have a £600 budget from the University of Bristol because I am completing a MSc by Research

6. **Does this source of funding place any restrictions on public dissemination (publication, etc.) of the results of the research? If yes, please say what these are.**
No.

7. Background and aims of the research:

It is becoming increasingly important to gain a full understanding of the public's perception of genetics and how best to communicate genetics to the public. Indeed, a top priority of genomics research is to cultivate communication strategies that will improve public understanding of genomics (McBride et al. 2010). The controversial nature of genetics means the involvement of the public is crucial to increase accountability in the decision-making process (Samuel & Farsides, 2017). Thus, the present research will explore public attitudes towards the integration of genetics into healthcare.

First, focus groups will be held to ask members of the public about their concerns and expectations towards the use of genetics in healthcare (study A). The aim is to uncover the necessary educational or additional information required to ensure individuals feel comfortable with the use of genomic medicine. Indeed, past researchers have recognised that attempts to understand public attitudes could uncover sources of scepticism and gaps in knowledge, which in turn can be used to guide communication efforts (Henneman et al., 2012).

Second, semi-structured one-on-one interviews with members of the public will also be conducted to establish the information they would consider relevant for making their decision to give their genomic data to the NHS (study B). Indeed, past research indicates a need to move away from imposing academic-focussed knowledge on the public and focus on supplying decision-relevant information (Condit, 2010). Similarly, Bates et al., (2005) argued that scientists should not treat the public as students who need facts but as conversational partners and found that a more influential form of public engagement is a presentation of not just *more* data, but data that is *more relevant* to the public.

The qualitative nature of these studies will allow for detailed, in depth responses that may open up complexities that had not previously been considered. Such rich responses are vital if we are to inform changes to practice based upon a full understanding of public perception of genetics. Further, the qualitative nature of the data may also help to highlight the role that personal experience plays in

an individual's acceptance of genomics. For example, Parsons & Atkinson (1992) found that participants' understandings of their chances of developing a genetically transmitted disorder were interpreted by translation into concepts, i.e. reproductive risk value, that were more personally meaningful. Similar, another study found that individuals with first-hand experience of genetic illness were simultaneously more optimistic about gene therapy technologies and more aware of the need for further conversations about the issues (Barns et al. 2000). Overall, it is recognised that members of the public display an impressive ability to draw on their existing knowledge and experience to understand difficult concepts (Davison, Barns & Schibechi, 1997), and this ability may be highlighted through the collection of rich, in-depth data.

Overall, it is recognised that an open dialogue is a necessity to ensure public trust with the aim to 'make genomics everyone's business' (Davies, 2017). Therefore, the goal of this research is to bring psychological science to genomic medicine in order to improve the communication approach of genomics in a way that expands public understanding and increases acceptance.

References

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Henneman, L., Vermeulen, E., Van El, C. G., Claassen, L., Timmermans, D. R., & Cornel, M. C. (2013). Public attitudes towards genetic testing revisited: comparing opinions between 2002 and 2010. *European Journal of Human Genetics*, *21*(8), 793.

McBride, C. M., Bowen, D., Brody, L. C., Condit, C. M., Croyle, R. T., Gwinn, M., & McLeroy, K. (2010). Future health applications of genomics: priorities for communication, behavioral, and social sciences research. *American journal of preventive medicine*, *38*(5), 556-565.

Parsons, E., & Atkinson, P. (1992). Lay constructions of genetic risk. Sociology of Health & Illness, 14, 437–455.

Samuel, G. N., & Farsides, B. (2017). Public trust and 'ethics review'as a commodity: the case of Genomics England Limited and the UK's 100,000 genomes project. *Medicine, Health Care and Philosophy*, 1-10.

8. Who will be recruited to participate in the research?

Adults (over 18 years of age), who are UK residents and have English as their first language or an equivalent level of fluency

9. How many participants will be recruited?

Study A: 2 focus groups with 8 participants each
Study B: 10 interviewees

10. How will the participants be recruited?

Advertisement on social media and email.

11. Are there any potential participants who will be excluded. If so, what are the exclusion criteria?

Yes. Exclusion criteria:

- Individuals under the age of 18
- Individuals who are not a UK resident
- Individuals who do not have English as their first language or an equivalent level of fluency

12. Where will the research take place?

University of Bristol, in any room within the Priory Road complex. The testing will be done in the evenings to allow ease of attendance for individuals with full-time jobs. A co-facilitator will be on site to provide support.

13. How will informed consent be obtained from all participants or their parents/guardians prior to individuals entering the research study?

Both study A & B:

Prior to the date of the study, participants will be sent the information sheet electronically, which will explain the nature, purpose and risks of the study to the participant. The email address of the lead investigator will be provided so participants may ask any questions prior to participation. When participants attend the study, they will be given a hard copy of the information sheet and a consent form to sign. There will be no time restriction on how long participants take to decide on whether to participate. Therefore, participants will be given sufficient time to read the information and consider any implications. Participants will be informed that they are free to withdraw at any time and will be encouraged to ask any questions they may have about the study.

14. Will the study involve actively deceiving the participants?

No.

15. Will participants be made aware they can drop out of the research study at any time without having to give a reason for doing so?

Yes.

16. Outline the design of the research study and list the procedures to which the participants will be subjected, the anticipated testing time and any treatments administered.

Two studies will be conducted and both will be recorded – please see below for details.

Study A: This is an exploratory study that will be delivered via 2 focus groups with 8 members of the UK public in each. Participants will be invited to attend a one-hour discussion group with 7 other members of the public, that will explore their concerns and expectations regarding the movement of genetics into healthcare. Semi-structured questions will be asked to promote discussion within the group. Demographic information will be collected at the start. A Dictaphone from the School of Psychological Science will be used to record the session. After the second focus has been held, within a week the data will be sent to University Transcriptions to be transcribed. After transcription, the original recordings will be destroyed.

Study B: An exploratory study through 10 one-on-one interviews with members of the UK public. Participants will be invited to attend a 1 hour discussion with a University of Bristol researcher (me). The interview will be semi-structured, with mostly open-ended questions. Demographic information will be collected at the start. A Dictaphone from the School of Psychological Science will be used to record the interviews. Immediately after each interview, a University of Bristol researcher (me) shall then transcribe each interview, before destroying each original recording.

17. Describe potential risks to participants (physical, psychological, legal, social) arising from these procedures.

None.

18. How will participants be debriefed?

At the end of both studies, I will verbally debrief the participants as well as giving them a hard copy of the debrief sheet.

19. Is any reimbursement of expenses or other payment to be made to participants?

Yes. Participants will be reimbursed for travel, refreshments will be provided during the studies and each participant will be given a voucher for £10 as a thank you for taking part.

20. Will personal data, beyond those recorded on the consent form, be used in the research?

Study A & B:

Basic demographic information, such as age, gender, ethnic group, and education level will be collected and used in the research. Also, participants will be asked whether they have children, whether they work for the NHS, when they were last taught about genetics, and whether they come across genetics in their workplace environment. This information will only be used to describe the sample – answers will not be linked to the individual's personal information.

21. Will the participants be audio-taped or video-taped?

Yes. Participants will be audio-taped.

22. When will this research be completed? (Give a date)

1st July 2019.

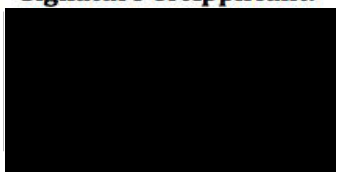
23. Any other relevant information

The documents attached in this ethics application are as follows:

1. Study A information sheet
2. Study A consent form
3. Study A debrief sheet
4. Study A personal data – this outlines the questions that will be asked of participants to indicate the demographic of the group
5. Study A recruitment advert – this advert will be used on social media and email to gather participants
6. Study A topic guide – this is an outline of the questions that will be asked by the researcher to the focus group during the study
7. Study B information sheet
8. Study B consent form
9. Study B debrief sheet
10. Study B personal data - this outlines the questions that will be asked of participants before the interview begins to provide information about the individual characteristics of each interviewee
11. Study B recruitment advert - this advert will be used on social media and email to gather participants

12. Study B topic guide - this is an outline of the questions that will be asked by the researcher to the interviewee during the study
13. Materials from the 100,000 Genomes Project – I have attached the information sheet and consent form for adults with a rare genetic condition and for their adult family members in the 100,000 Genomes Project. These will be given to the interviewees to gain feedback about the process of consent.
14. References of the study outline

Signature of Applicant: **Date:** 7.11.18



Conditional letter from Faculty of Science Human Research Ethics Committee.

School of Psychological Science
12a Priory Road
Bristol BS8 1TU
Telephone: (0117) 928 9000

21st February 2019

Miss Helena Davies
School of Psychological Sciences

Dear Miss Davies,

Ref: 77803
Thoughts from the UK public on the use of genetics in healthcare: Concerns, expectations, and the provision of decision-relevant information

The ethics committee have considered the above proposal which has received a conditional opinion requiring the following required changes to be seen by the chair of the committee:

- The committee requested in the inclusion criteria that only adults are being recruited.
- The committee noted that the study referred to taking place in a particular room within the Priory Road complex and thought it might allow for more flexibility if the application states that any room in Priory Road complex will be used to avoid needing an amendment just to interview in another room.
- The committee requested clarification that a risk assessment has been conducted.
- The committee noted that in the study documentation sometimes the time taken is said to be 30-40 minutes, sometimes it is mentioned to be 1 hour.
- The committee noted the monetary compensation for study participation and advised that generally speaking the rate for study participation is £10 a hour.
- The committee advised that when questions regarding gender are asked it is preferable to have the question as a free text box for participants to write in what they identify as rather than a choice of particular options.
- The applicants don't mention how long audio-recordings will be kept (i.e., when audio-recordings will be transcribed, and if/when the audio-recordings will be destroyed).
- The committee observed that the researcher doesn't appear to mention what device will be used to record interviews and recommended something that is encrypted and secure for vulnerable data.
- The committee noted the PIS mentions Faculty of "Life Science" instead of "Psychological Science" Research Ethics Committee and wants this to be updated to reflect the new faculty structure at the university and structure of ethics committees accordingly.
- The committee noted the distinction between study A and study B and wanted it clarified that both studies are being recorded and whether there is a final consent form for study A as well as study B as it currently appears it may only be for study B.

Our response to the Faculty of Science Human Research Ethics Committee.

21st February 2019

Dr. Jonathan Evans
Chair- School of Psychological Science Research Ethics Committee

Dear Dr. Evans,

Thank you for your response regarding my ethics application (**Ref: 77803**) **Thoughts from the UK public on the use of genetics in healthcare: Concerns, expectations, and the provision of decision-relevant information.**

Below I have outlined the required changes and how I have addressed them. Please see attached for the amended documents – all changes have been tracked.

- *The committee requested in the inclusion criteria that only adults are being recruited.*

Addressed: In the 'Full ethics' document, I have amended question 8 regarding who will be recruited to participate, so that it reads 'Adults (over 18 years of age), who are UK residents and have English as their first language or an equivalent level of fluency'.

- *The committee noted that the study referred to taking place in a particular room within the Priory Road complex and thought it might allow for more flexibility if the application states that any room in Priory Road complex will be used to avoid needing an amendment just to interview in another room.*

Addressed: : In the 'Full ethics' document, I have amended question 12 regarding where the research will take place, so that it reads 'University of Bristol, in any room within the Priory Road complex'.

- *The committee requested clarification that a risk assessment has been conducted.*

Addressed: I have attached the out of hours risk assessment for Study A which has been submitted and approved by the School of Psychological Science. I have also attached the risk assessment for Study B. This is yet to be sent to the School of Psychological Science as I am conducting Study A first.

- *The committee noted that in the study documentation sometimes the time taken is said to be 30-40 minutes, sometimes it is mentioned to be 1 hour.*

Addressed: I have made the necessary changes in the Study B documents, so that it now indicates the study will take 1 hour.

- *The committee noted the monetary compensation for study participation and advised that generally speaking the rate for study participation is £10 a hour.*

Addressed: I have made the necessary changes in both Study A and Study B documents, so that it is now clear that the participants will be reimbursed £10 for their time.

- *The committee advised that when questions regarding gender are asked it is preferable to have the question as a free text box for participants to write in what they identify as rather than a choice of particular options.*

Favourable opinion letter from the Faculty of Science Human Research Ethics Committee.

University of BRISTOL

SCHOOL OF PSYCHOLOGICAL SCIENCES
12a Priory Road
Bristol BS8 1TU
Telephone: (0117) 928 9000

9th April 2019

Miss Helena Davies
School of Psychological Sciences

Dear Helena,

Ref: 77803
Title: Amendment 1 - Thoughts from the UK public on the use of genetics in healthcare: Concerns, expectations, and the provision of decision-relevant information

Thank you for providing the following changes to your ethics proposal as detailed in your email dated 02.04.19:

These changes have been reviewed by the Chair of the ethics committee and approved. Your ethics approval code remains **31011977803.**

Good luck with the continuation of your study.

Nathan Street
Research Governance Administrator

pp
Dr. Jonathan Evans
Chair- School of Psychological Sciences Human Research Ethics Committee

Appendix 5 Question guide for the focus groups.

- Would you have your genetic data tested by the NHS if you were at risk for a single gene (monogenic disorder)?

- Would you have your genetic data tested by the NHS if you were trying for a baby and wanted to know your carrier status?

- Would you have your genetic data tested by the NHS you were at risk of carrying a high penetrance mutation?

- Would you have your genome sequenced by the NHS if you were told that knowledge of your genetic data may improve the effectiveness of a medical intervention that you required?

- Would you want to know any information about your genetic sequence other than the specific genetic variant under investigation?

Do you agree or disagree with the following statements:

- 'I am optimistic about the possibility of medical discoveries as a result of genetic research'

- 'I expect **medical professionals** to consult me if a genetically related family member is having a genetic test'

- 'I expect **genetically related family members** to consult me if they are having a genetic test'

- 'I would be willing to share my anonymised **patient record** for medical research purposes'

- 'I would be willing to share my anonymised **genetic data** for medical research purposes'

- 'I feel suspicious about genetic studies for the improvement of health: hidden political or economic agendas may be behind them'

- 'I would like to hear more information from scientists and healthcare professionals about genomic medicine'

What do you agree with *most?*

- 'The use of genetic data to inform medical research is **overly intrusive**' OR 'The use of genetic data to inform medical research is **not any more intrusive than using any other data**'

- 'I would be more concerned about my **genetic data** being leaked than my **credit card information'** OR **'I would be more concerned about my credit card information being leaked than my genetic data'**

- 'For me, DNA information is **the same** as any other medical information' OR 'For me, DNA information **is different** to other medical information'

Imagine yourself in the following scenario…

- *You have an opportunity to have a genetic test that may reveal that you are going to develop a condition that currently does not have any medical treatment options…* What do you agreee with most?

Knowledge is power: I would rather have the genetic test, so that I can prepare financially, emotionally, and practically if I am found to have a genetic variant that will lead to a (currently) incurable disease OR **Ignorance is bliss**: I would rather not have the genetic test, because if I am found to have a genetic variant that will lead to a (currently) incurable disease, I would worry about it too much to be able to carry on enjoying my life to the full

Finish this sentence:

- 'I am most **concerned** about…'

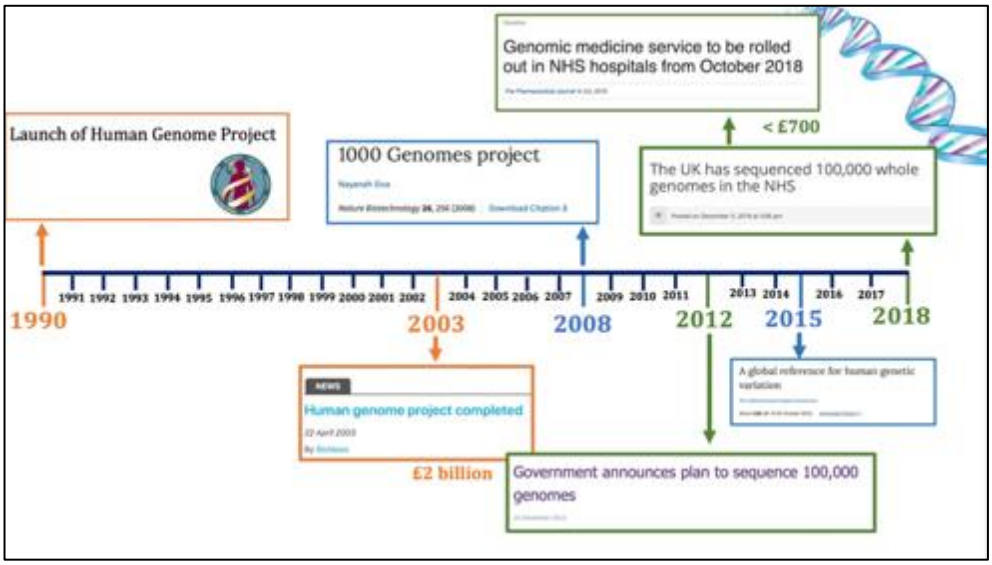
- 'I am most **optimistic** about…'

Appendix 6 Presentation slides for the qualitative study.

Slide 1:

Video* (0:28 – 6:21)*: https://www.youtube.com/watch?v=IXamRS85hXU

Slide 2:



Slide 3:

Genetics in healthcare: What is the current situation?

- Single gene disorders
  Huntington's, polycystic kidney disease, hypercholesterolemia
- Cancer
  Melanoma (skin cancer), leukaemia, colon, brain and breast cancers
- High penetrance mutations
  BRCA1, BRCA2, APOE

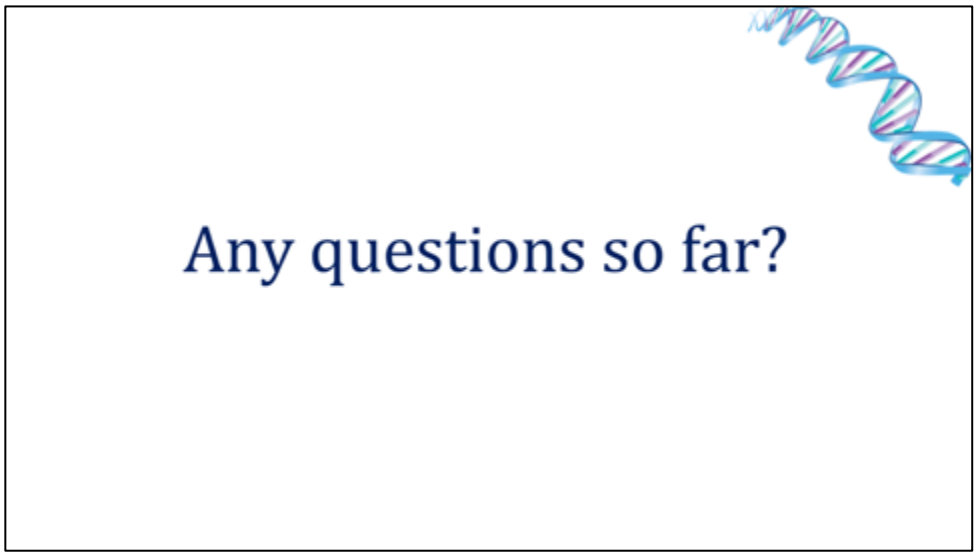
Slide 4:

Genetics in healthcare: The future?

One-size-fits-all → Personalised medicine?

'The foundations for this step change in healthcare are already being put in place.'
NHS England

Slide 5:



Appendix 7 Coding tree for qualitative study.

Detrimental psychological impact of results
Anxiety for self
Anxiety for family members
Hypochondria
"Waiting for it to happen"
Power of the mind
Disclosure and discrimination
Importance of anonymity
Discrimination from organisations (insurance, employers)
Status as a romantic partner
Crime & the police
Family planning
Preparation – informed decision making
Remaining uninformed
Pressure to *not* start family
Charting possible futures
Genetic modification
New ideas & suggestions for the future system of genomics
Genetic exceptionalism: "DNA is different"
Genome linked to identity
"Playing God"
Lack of privacy
Intrusiveness of genetic data
Knowledge is power
Tailored treatment
Mental preparedness
Cherishing the time that you have
Practical preparation
Effective preparation for illness
The 'greater good'