ETHICAL ISSUES RELATING TO INVOLVEMENT OF CANCER PATIENTS IN THE 100,000 GENOMES PROJECT

Qualitative Research Findings

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1 Executive summary

Background and method

Research was commissioned to support and inform Genomics England’s 100,000 Genomes Project. The key objective of this research was to explore the views of patients with a diagnosis of cancer and their carers on the ethical issues relating to involvement in the 100,000 Genomes Project. The specific ethical issues explored were:

- Commercialisation
- Data access and privacy
- Perpetuity
- Feedback and re-contact
- Non-specific uses

Additionally, the research aimed to identify preferences in terms of data access and feedback.

Four qualitative focus groups were carried out, each involving six participants and lasting for 90 minutes. Groups were conducted in Manchester and Birmingham. The sample included those who have had a diagnosis of cancer and those who care for others with a diagnosis of cancer. Each group included a spread of age groups, social grades and cancer types.

Key findings

General attitudes towards medical research

The research identified a range of attitudes towards medical research. These spontaneous attitudes and concerns provided an important backdrop to the wider ethical questions, as it is possible, by implication, that these will feature in potential participants’ decision-making around whether or not to volunteer to take part in Genomics England’s work. Participants commonly associated medical research with drugs trials, and it would be helpful to make clear the distinction between 100,000 Genomes Project and drugs trials, so participants understand what would be involved.

Responses to Genomics England concept

To provide context to the discussion of ethical issues, research participants were asked for their general views on the 100,000 Genomes Project. As it was anticipated that research participants may not have a great deal of knowledge of this, information was provided to prompt discussion (see Appendix 7.3). It was felt, above all, that this type of scientific work is important and has a great potential to prevent and cure disease, thus saving lives. Participants’ spontaneously associated positive benefits with the project, relating to the furthering of medical understanding and contributing towards the ‘greater good’. The knowledge base that the project would potentially generate was thought to be invaluable to help protect against
disease, as genetic understanding was seen as key to curing cancer – a disease recognised as affecting a high proportion of people.

Respondents also spontaneously raised concerns about the project, and privacy and confidentiality was by far the main concern for most participants. This related to the protection of information about their personal identity, which they did not wish to be linked directly to their genome data. However, Genomics England were generally trusted in having the volunteers’ best interests at heart, and would safeguard confidentiality as much as could be reasonable done.

**Ethical Issues**

Participants were not explicitly asked to rank the ethical issues in order of priority. However, a clear hierarchy of relative perceived importance of the various ethical issues emerged from the discussions. Responses to each of the ethical issues that were explored are addressed in order of importance to participants.

**Data access and privacy**

Data access and privacy was a top of mind concern for almost all participants. Key concerns related to the potential for data to be accessed by insurance and other financial services companies, with the belief that there could be a risk of personal detriment if these types of organisations had access. The protection of participants’ personal identity was also regarded as a basic requirement for involvement in the project, and any prospective decision to take part was seen as contingent on this. Participants were happy for medical and lifestyle information to be linked with their genome data, but there was a clear desire to have this separated from personal identifying information.

**Feedback and re-contact**

There was a wide variation in participants’ views regarding individual feedback, with some participants saying they would want to know as much as possible about the findings relating to their genome data, whereas others thought they would prefer to know nothing at all. The drivers behind the desire for individual feedback related to the opportunity to make different choices in life such as lifestyle choices and choices about having children. Conversely, the barriers to participants’ wish to receive individual feedback very much centred around a desire to not know about potential conditions.

Participants generally felt there was a need to be able to select feedback and re-contact options on an individual basis, in order to reflect each individual’s personal circumstances and preferences. Most participants expected that there would be a form of ‘contract’ setting out such options at the outset.

In terms of potential withdrawal options, most participants felt that it would not be fair to impact upon the project as a whole by requiring that existing information and samples be destroyed altogether if a volunteer wishes to withdraw. However, there was a desire for an option to be available, if requested, to walk away and avoid any further personal contact, and/or to de-link data from personal medical information.
Most participants felt that the obligation be informed of other trials relevant to them was acceptable, as this was not an offer that needed to be taken up.

**Commercialisation: potential for personal impact**

In the course of discussion of the ethical considerations around commercialisation, two broad themes emerged. The first of these themes focused on the potential for personal impact as a result of participation in the 100,000 Genomes Project, and was very much allied to the issues relating to data access and privacy. The second theme revolved around the profit-making agenda of the commercial companies who would be involved.

The key concerns for participants relating to commercialisation focused upon the potential personal impact it could cause and a fear of their data being ‘re-shared’ or ‘sold on’ to third party companies who would seek commercial advantage as a result. There was a fear that participants themselves, or their young families, would be vulnerable to discriminatory policies from ‘real world’ companies such as insurers and banks. This was felt to be potentially detrimental to future life chances. However, there was a general assumption amongst participants that the intention would be that information relating to volunteers’ personal identity would remain confidential.

**Commercialisation: involvement of profit-making**

Although a lesser issue for participants, the involvement of profit-making companies in the project potentially becomes a key concern for some. There was a degree of scepticism regarding the motivations of commercial companies, who would be driven by the demands of their shareholders. Having said this, the scepticism expressed was set alongside a clear realisation and pragmatism that commercial companies would be in the best position to make actionable developments from the research data, and that they work in partnership with the NHS or government departments already. Inevitably, they would profit as a result, but the benefit would be faster medical advancements.

**Perpetuity**

Access to data in perpetuity was a lesser issue for most participants, who appeared to see assumed ongoing consent as the expected default position for involvement in the project. The commonly held view was that ‘if you’re in, you’re in’, and the data would be retained and used for as long as it was useful. One key proviso was made in relation to this point, however: that if a strong reason to want to withdraw from the project should present itself, there should be provision in place to accommodate this. Some participants referred to extreme scenarios and the impact they would be likely to have on their level of trust – particularly breaches to security and personal privacy. Participants envisaged that, as part of an initial contractual agreement to take part in the project, there would be a structured process for withdrawal in place. This would most likely not be used, as they saw it, but would provide a necessary fallback and as such, was regarded as a point of good practice.

**Non-specific uses**
There was very little concern expressed regarding non-specific uses of data, provided these were legitimate medical research uses. Indeed, most participants anticipated that there would be an ‘all or nothing’ commitment level to the project. Donation to the project was viewed as taking part in a fight or ‘war’ against disease at a very general level, and for the ‘greater good’ – for medical benefit to anyone, anywhere. Personal benefits were not expected from the project, and there was no sense of an implicit trade-off being made between donation of data and direct benefit to the individual or the individual’s family. Several participants likened this to blood donation; you ‘do your bit’ by giving blood, without knowing who will benefit from your donation. For this reason, non-specific uses were regarded as not only inevitable, but as a positive feature of the project.

2 Introduction

2.1 Background

Genomics England was set up by the Department of Health to deliver the 100,000 Genomes Project. By 2017, the project will sequence 100,000 genomes from patients with rare disease or cancer. Genome data will be linked with medical records and studied by doctors and researchers, including those from industry. For cancer it will mean greater insight into its cause, progression and most effective treatment. Some participants with cancer may benefit personally but it is more likely that participants will benefit people like them in the near future through new medicines, treatments and diagnostics. Undertaking this very large project will also make it possible for the NHS to have a genomic medicine service by 2018, so that any sick person who needs it will be able to use it in the future.

There are some important ethical issues relating to participants’ data; who should have access to it and under what circumstances; and what participants would like to be told about the findings.

Genomics England appointed independent research agency GfK NOP to speak to potential volunteers to understand their perceptions of ethical issues related to the 100,000 Genomes Project.

2.2 Objectives

The key objective of this research was to explore the views of patients with a diagnosis of cancer and their carers on the ethical issues relating to involvement in the 100,000 Genomes Project. The specific ethical issues explored were:

- Commercialisation
- Data access and privacy
- Perpetuity
- Feedback and re-contact
- Non-specific uses
Additionally, the research aimed to identify preferences in terms of data access and feedback.

2.3 Research approach

2.3.1 Method

Four qualitative focus groups were conducted, each involving six participants. Each group lasted for 90 minutes to allow for in-depth exploration and discussion of all of the ethical considerations.

It was anticipated that awareness and understanding of the 100,000 Genomes Project was likely to be low, and this was found to be the case. For this reason, a briefing exercise was used where respondents were shown and read out a series of show cards, giving descriptions of Genomics England and the 100,000 Genomes Project (see Appendix 7.3.). This ensured that all respondents had a similar understanding prior to discussion of the ethical implications of the project.

During the groups, respondents were set barometer exercises following discussion of each ethical issue. This allowed for individual feedback and reflection on the ethical issue discussed. An example barometer exercise is provided in Appendix 7.4.

2.3.2 Recruitment

The research participants were recruited via a free-find method. This involved recruiters identifying potential participants in their local area by a variety of means including controlled snowballing (asking those interviewed whether they know anyone meeting the criteria) and contacting people via local organisations and support groups.

A screening questionnaire (see Appendix 7.1) was developed, with pre-qualification criteria to ensure that the research participants met required sampling criteria, and that anyone with specialist knowledge of the subject could be excluded. For example, potential participants were not included if they worked within the medical or pharmaceutical professions, as their views of the project may be somewhat different.

Research was conducted across two locations: Birmingham and Manchester. The focus groups took place in urban centres, with participants travelling to them from surrounding areas, in order to ensure a mix of those living in urban, suburban and more rural areas. Due to the nature of the study, each group was over-recruited by two participants to ensure maximum participation in the research.

2.3.3 Sample

Four focus groups were conducted in total. The sample included both those who had themselves been diagnosed with cancer, and those who care for others with a diagnosis of cancer. A range of cancer types was represented in each group, to
include all of the most common cancer types as well as some less common cancers. Each group included those from a spread of age groups and social grades.

A breakdown of the sample composition can be viewed in the table below.

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<thead>
<tr>
<th>Carers</th>
<th>Birmingham</th>
<th>Manchester</th>
</tr>
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<tbody>
<tr>
<td>Group 1</td>
<td>Male</td>
<td>Group 3</td>
</tr>
<tr>
<td>Group 2</td>
<td>Female</td>
<td>Group 4</td>
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The carer groups included those who cared for:
- Immediate family (spouses, children);
- Others (parents, siblings, other relatives, neighbours).

2.3.4 Sample characteristics

Some differences were identified in the mindset and outlook of the research participants, based on demographic characteristics, and carer relationships. These differences had an impact on participants’ views of the issues discussed.

Amongst carer groups there was a mix of those caring for immediate family, and those caring for others such as other relatives or neighbours. This range was reflected in varying intensity of feeling on the ethical issues discussed.

It was also found that there was a differing outlook amongst male and female patients:
- Male patients were generally more cynical about the involvement in the project of profit-making organisations;
- Male patients appeared generally a bit more negative about their diagnosis and about the medical profession;
- Those with less trust, or a poorer relationship with their GP or health professional, generally had a slightly less positive outlook on medical research.
3 General attitudes towards medical research

3.1 Spontaneous understanding of medical studies, projects and trials

General attitudes towards medical research were explored, to provide context for the perceptions of Genomics England and the 100,000 genomes project. It was notable that spontaneous responses from participants were related to drugs trials, including those for which participants are paid, rather than sample- or data-based studies.

“I signed up – I never went – to one called ‘Flu Camp. They’re always advertising now, on the radio and stuff, and it’s basically to study the effects of flu on people with different illnesses or on different medications...It’s [the motivation is] financial – I mean, where else can you get £3,500 for sitting on your bum for 10 days?”

Male patient, Manchester

On further discussion, however, it emerged that several participants in fact had personal experience of the latter type of study, but it appeared that often these had been put out of participants’ minds since, and so had not been top of mind.

3.2 Spontaneous views on benefits of medical research

Participants spontaneously cited several benefits of medical research.

It was viewed as essential to test new drugs, and on humans rather than on animals, in order to evaluate their effects, move forward with technologies and advance medical science.

“I’m all in favour of them if people are willing to do it. I mean, they’ve got to find out things, at the end of the day – otherwise probably a lot of us wouldn’t be here. So I’m in favour of drug trials. I’d be a bit scared to do one myself, to be honest... But I’m in favour generally.”

Female patient, Birmingham

Participation in medical research was seen as representing a potential means to access and opportunity to benefit from new, potentially life-saving drugs before they are widely available to patients.

“The time it takes to get whatever results have come out of it available to people who are ill, you know - we’ve come up with this wonder drug, but it’ll take 10 years to prove that it’s OK. And I think...if you’re desperate, you want it. You won’t be here in 10 years!”

Male patient, Manchester

Where chances of survival are otherwise low, participants’ view was that you may as well take part in a trial, as you have nothing to lose and, if nothing else, you are helping others.
Several participants spoke of being very conscious of having benefited from the experience of patients who have gone before them, whose past suffering had contributed to advances in medical knowledge.

“You might do it for purely altruistic reasons – you know, you might think you’re helping people. One thing was clear to me, when I had my diagnosis and they said bang, bang, bang, bang - ‘This is what’s going to happen’ – well, they only knew that because some other poor bugger’s been before you... So you can only move forward from experience, and if part of that experience is trials like this, OK.”

Male patient, Manchester

3.3 Spontaneous views on concerns relating to medical research

Participants also spontaneously cited several concerns relating to medical research.

It was seen as intrinsic to the trialling of new drugs and treatments that there would be an element of risk. Anecdotes were shared about trials that had featured in the news, because of damage caused to people taking part.

“I used to work [as painter/decorator] in hospitals, and the one at Leeds, it was one of these medical places where people were paid to go for a week. I can’t remember what they were doing to them now, but three of them died – they turned blue, these people, it was all in the papers... Yeah, they put them up for a week or whatever and they do all these trials.”

Male patient, Manchester

Given this element of risk, concerns were expressed about the motivation of volunteers, who might be too much tempted to take part in such trials to earn financial reward, rather than for medical or altruistic reasons.

“I’m in favour, but what would concern me is the screening of the people that go in for the drug trials, because you get paid…and they they go off, you know, because they need the money... That would be a concern of mine... – that they were doing it for monetary recompense rather than moral recompense.”

Female patient, Birmingham

A few participants felt that the balance of emphasis and resources was tipped in favour of trialling drugs, rather than more ‘natural’ forms of treatment. This was seen as a consequence of the fact that drugs companies exist in order to make profits for their shareholders.
“Well I was looking on the internet last night, and they reckon that drugs companies pay doctors to give drugs, and then natural things are suppressed which can be safer. And you just don’t know what to do. The fear of cancer sends you down the drugs line. You know, if it was an even balance it would be better.”

Female patient, Birmingham

Spontaneous mention was made by one participant of the failure of a previous research study to offer personal feedback on findings to participants, which she viewed as strange and unfair.

“Many years ago, my cousin, she did a study – every year she went for blood tests. It was for breast cancer...but what they did say to her was that, even if they picked anything up, they wouldn’t tell her. And I found that quite strange, that they would be doing this research but if they found anything, they wouldn’t say ‘Actually, you need to go to your GP’... And to be honest, she has developed breast cancer, so I do find that research quite strange really... It was very wide, this research, and yet they did say that they wouldn’t be notifying people if anything did come up in the blood. I found that quite strange.”

Female patient, Birmingham

A few participants – particularly men whose wives had taken part in drugs trials, when they were very ill indeed - felt that it was difficult in practice to have to take part and not know whether or not you were among the number being given the treatment in question, as opposed to a placebo.

“I think the only one [concern] I would have is, I think, if there was a placebo drug and a real drug...I think ‘Give me the drug, or don’t give me the drug - don’t pretend I’ve got something’.”

Male carer, Birmingham

3.4 Implications for Genomics England

These spontaneous attitudes and concerns relating to medical research generally provide an important backdrop to the project, as it is possible, by implication, that these will feature in potential participants’ decision whether or not to volunteer to take part in Genomics England’s work. The key factors are as follows:

- Since there appear to be generally more personal and vested interests related to drugs trials, it would be helpful to make clear immediately the distinction between them and 100,000 Genomes Project in potential participants’ minds;
- Willingness to participate and to give consent seems to be potentially less of an issue for sample and data-based trials, particularly if what is involved is closer to one-off donation than an ongoing programme of appointments;
• There seems to be a strong motivation amongst participants to help advance medical knowledge, in spite of the sensitivities also voiced surrounding the commercial aspects of trials;

• It seems likely that some participants at least would want to receive feedback if anything significant were picked up on the basis of their own genome sample.
4 Responses to Genomics England concept

To provide context to the ethical discussion, research participants were asked for their general views on the 100,000 Genomes Project. As it was anticipated that research participants may not have a great deal of knowledge of this, information was provided to prompt discussion (see Appendix 7.3).

4.1 General responses

While there was a very general awareness of DNA and genetics, this was as much in relation to the contexts of crime detection (e.g. DNA testing of suspects by the police) and genealogy (e.g. the recent discovery of the remains of King Richard III) as to the context of hereditary disease and gene-based or stem cell therapies.

There was very little recognition and awareness of genomics specifically. Although the work in this area was regarded as important, interesting and clever, most participants felt the subject matter to be hard to relate to.

“It’s been on the news and in the papers. It’s all very high level stuff but, to be honest, if it went any deeper it would just lose me anyway. I mean, I know what DNA is.”

Male patient, Manchester

Although some felt the subject matter to be rather remote and abstract – with comparisons being made with the Hadron Collider and science fiction-style films - there was a certain level of curiosity about what has happened since the headlines about the Human Genome Project in 2003.

“Yes I remember when that became common knowledge, but what’s happened since [2003]. I don’t know...I just remember that it was called the genome, and they could look at it and know people’s, the set-up of their body.”

Male carer, Birmingham

There was also extremely limited recognition and awareness of Genomics England and of the 100,000 Genomes Project and related scientific work. Participants did express positive interest and curiosity, however – with questions about what lies behind the project objectives, for example.

“I’m intrigued as to why 100,000 is the magic figure. Is that because that’s what’s manageable, or has somebody worked out that 100,000 sets of information will give a complete picture?”

Male patient, Manchester

It was felt, above all, that this type of scientific work is important and has a great potential to prevent and cure disease, thus saving lives.
“I’m quite happy that there are people clever enough to understand it, and to do this kind of work. It’s advancing human knowledge – that in itself is a good thing.”

Male patient, Manchester

Two of the research participants were aware of Genomics England - one through a family member who was formerly a health care professional, and another through recent news headlines. One other participant was aware of a data storage facility in Manchester, which he had seen featured in the news.

“I know about it from recently, because they’re trying to do 100,000 people, so they can find out what diseases, and how it effects the genome. Genetics is the way to cure cancer...It was on the news. My wife, she used to be a nurse, so she does a lot of research and I just sit there and listen. I do think genetics is the way forward.”

Male carer, Birmingham

Broadly, Genomics England and their work was seen as trusted, and as having the potential to save lives. However, concerns were expressed by a few participants about the potential for new scientific discoveries to be abused or used for destructive purposes, ranging from ‘designer babies’ to warfare.

“This [the Project information] is extremely uplifting, and very positive and forward-looking, and I don’t think you can probably have any kind of issues with that. But in anything like this there has to be a flip side, maybe a darker side...What happens if it’s then sold to a third party? What happens if it somehow finds its way to the military? Could this information be used to develop, I don’t know, chemical warfare weapons and things?”

Male patient, Manchester

In addition, a small number of comments were made about this type of work being seen as ‘macabre’ or as ‘delving too deep’, suggesting that the subject matter was found by a few to be philosophically challenging.
4.2 Comparing 100,000 Genomes Project to other forms of medical research

Participants cited differences when comparing the 100,000 Genomes Project to other forms of medical research. These differences are summarised in the table below:

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<th>Timescale</th>
<th>Medical Research (e.g. drug trial)</th>
<th>100,000 Genome Project</th>
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<tr>
<td>Short-term benefit</td>
<td>Receiving ‘treatment’</td>
<td>Long-term realisation</td>
</tr>
<tr>
<td>Receiving ‘treatment’</td>
<td>A demanding process, active involvement</td>
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<tr>
<td>A demanding process, active involvement</td>
<td>Potential for direct benefit for individual or own charity / condition</td>
<td></td>
</tr>
<tr>
<td>Potential for direct benefit for individual or own charity / condition</td>
<td>The ‘greater good’: improved health &amp; fight against disease</td>
<td></td>
</tr>
<tr>
<td>Patient</td>
<td>Citizen / volunteer</td>
<td></td>
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It is notable from these differences that participants’ intuitive understanding was that the donation of a person’s genome to the 100,000 Genomes Project would be primarily for longer-term benefit to medical science generally, rather than for their own shorter term benefit as individuals.

4.3 Spontaneous views on positives

Participants spontaneously associated positive benefits with the project, relating to the furthering of medical understanding and contributing towards the ‘greater good’. The potential knowledge that the project would generate was viewed as invaluable to help protect against disease, as genetic understanding was seen as key to curing cancer – a disease recognised as affecting a high proportion of people.

“Well my wife worries now about our daughter...So I mean, I think everybody – most people know someone who’s been touched by cancer. It’s one in three people or something. And I think most of us have probably said at some point ‘I wish that they could find a cure for cancer’...So I think anything that can help...
think people who...have got cancer or had cancer...if they could help going forward, I’m sure they would.”

Male carer, Birmingham

There was recognition that patients benefit today from the experience gained by the medical profession from the illnesses of others who have gone before. Implicitly, it was felt, it was natural to want to contribute in their turn to that ongoing development of knowledge.

“As we all understand, the poor souls that we’ve lost in the past to get us to this table today...If it wasn’t for those before us...So yeah, I’d have no hesitation in doing it.”

Male carer, Birmingham

Some participants also expressed the view that it is a good thing if health risks can be identified, so that pre-emptive action could be taken if necessary. This was felt to be the case particularly for the younger generation, with most of their lives ahead of them. Angelina Jolie was cited as an example of a well-known figure who was known to have taken such pre-emptive action.

The briefing materials (see Appendix 7.3) used in the groups referenced an example of HER2, to give participants a concrete understanding of how genetic data is used. There was immediate recognition of this example, and participants spontaneously related other personal experiences of gene tested treatment.

“My mum and dad have had lung cancer, and my mum was put on Tarceva because she had a gene – it’s brilliant, it is. But my dad hadn’t got that gene for him to be put on that – you’ve got to be at a certain threshold, haven’t you. So for my mother it’s perfect - you know, though it sent her like a zombie, it really did...as soon as she came off the tablets, she was running down the road. It was amazing. And it put her into remission.”

Male carer, Birmingham

The backing of Government and NHS was also viewed positively, as it was felt to be inclusive and less immediately associated with commercially vested interests.

There was no particular concern about the prospect of the donation of genome data, nor about such data being linked to anonymised lifestyle and medical information. However, there was a strong desire from most that data should not be linked directly to personal identity information.

“All it takes is for someone with a memory stick to lose it, and for someone else to find it. It’s been done before...people have left laptops with sensitive information on.”

Male patient, Manchester

4.4 Spontaneous views and concerns
Respondents spontaneously raised concerns about the project, relating to privacy and confidentiality, commercialisation, feedback and outcomes, and participation.

Privacy and confidentiality was by far the main concern for most participants. This related to the protection of information about their personal identity, which they did not wish to be linked directly to their genome data.

“I think it depends who’s got that information and, you know, who can access it. It’s like at the moment, the GP records and everything – various ones can get into that that really shouldn’t be able to... You know, if I drop down and the ambulance comes and takes me, they’ve just got to put my name in and it [my medical record] comes up. But who else can see that?”

Female carer, Manchester

In terms of commercialisation, spontaneous concerns were raised by some participants who felt cynical about the profit-making motivations of commercial drug companies who they anticipated would be involved with the project.

“There will be some large drugs firm involved in this, won’t there – some large drugs firm, because of the treatment that may be necessary... [That’s] not really very good, because it’s all profit. They might show sympathy, but the main thing is they’ve got to show their shareholders profit. I’m not very pleased about that really. I know it’s through the NHS, but still.”

Male patient, Manchester

Concerns were also raised about the risk that research results would not come fully to fruition if drugs were not developed as a result that it was economically viable ultimately to make available to patients.

“And then unfortunately they bring this new drug out – wonderful, wonderful – and then they can’t afford to give it to people. I mean, it just happened last week, didn’t it – the drug that gives you six months on breast cancer. They can’t afford to give it.”

Female carer, Manchester

A further concern related to commercialisation was the potential passing on of information to other profit-making third parties, such as insurance companies. Apart from the potential for breach of confidentiality, participants felt that if this type of organisation had access to their genome data, it could give rise to discriminatory policies which would impede on their own or their families’ future life chances.

“I think there can be some negatives, I’m sorry to say, because I think for some people, they may be the sort that would constantly be worrying about what’s going to happen... There are things that have been in the press about, you know, if you do know everything about that person, if they are more prone to getting cancer or an illness, then insurance can be a big problem.”
Opinion was divided regarding feedback and research outcomes. Some participants certainly felt that the current treatment of cancer already benefited from the progression of genomic research since 2003.

“That is surely – from taking samples from my tumour, that’s how they discovered I wouldn’t respond to Herceptin, because I hadn’t got that HER2 gene. I mean, that must be part of something they found out since 2003... But certainly that’s, I feel, helped in my treatment.”

Others, however, felt that there was a likelihood that knowledge about a volunteer’s genome might well create concern – possibly unnecessarily - around the propensity for disease.

“I read such a lot on people with breast cancer and their daughters... They have elective mastectomies at a very young age, and I think it’s all to do with the genetics, and probably the genomics... It’s not 100% that they’re going to get cancer, and yet they have everything removed, then they have reconstructive surgery, and then they may not have got it anyway.”

Another concern spontaneously expressed related to the potential for duplication by the 100,000 Genomes Project of existing research programmes currently being undertaken by cancer charities – organisations to whom participants are donating money.

“My concern is, we’ve got all these cancer charities...so are they not duplicating the work that cancer charities are doing? I mean, it’s brilliant that the government want to do something, but surely they should be working hand in hand with the charities... I feel as though, you know I give my money to Marie Curie, Cancer Research...all of these things, I give my money to that, they’re researching, researching, researching. So is it not doing the same thing?”

Finally, some participants raised queries and concerns regarding participation, as they felt it was not clear to them what level of involvement in the project – whether transient or ongoing over time - would be required of people who volunteered to take part. It was felt that the level of commitment required would need to be clear upfront, as it may be a critical factor in whether or not cancer patients would take part in the project. Due to existing commitments to attend hospital appointments, participants might find a need for ongoing involvement in the study overwhelming.

“If the study could be done in conjunction with treatment...because you spend enough time in hospital as it is, in my opinion. I spend 5-6 hours one week, and the
day before, blood tests a couple of hours, and then the next week – you’re going to be spending most of your life in hospital! I know it’s for the advancement of, but there’s got to be some happy medium between the two – them gaining the knowledge, but also you retaining some of your life.”

Male patient, Manchester
5 Ethical Issues

5.1 Hierarchy of perceived importance of issues

Participants were not explicitly asked to rank the ethical issues in order of priority. However, a clear hierarchy of relative perceived importance of the various ethical issues emerged from the discussions. This ranking is summarised in the table below, showing a progression from participants’ key concerns at the top down to lesser issues and concerns at the bottom.

* Commercialisation – potential for personal impact: If access is granted to commercial organisations such as insurance companies, this could result in negative impact on future life chances

** Perpetuity: A lesser issue unless a strong reason to withdraw arises later.

As is indicated on this table, the most strongly felt concerns revolved around the potential for personal impact or detriment on the lives of participants and their families, both in the present and in the future. Participants were rather less concerned about the uses to which donated information was put, and by whom, provided these were for legitimate medical purposes, and provided data relating to participants’ personal identity remained secure.

Responses to each of the ethical issues that were explored are addressed in detail in the sections immediately following.
5.2 Data access and privacy

Data access and privacy was very much top of mind as a concern for almost all participants, and was a topic raised spontaneously before any prompting on the specific ethical issues – as reported earlier in this report (see section 4.4 Spontaneous views on concerns).

The key concern expressed relates to the potential for data to be accessed by insurance and other financial services companies, with the belief that there could be a risk of personal detriment if these types of organisations had access.

“[They don’t need to know who you are, do they? They don’t need to know who you are. They need to know age and, you know, the area of where you live, because some of it [risk factors for disease] can be environmental – generic stuff.]”

Female carer, Manchester

This was a clear concern to participants in relation to their own privacy; it also extended beyond themselves as individuals to include the privacy of their families – particularly young children with the bulk of their lives still ahead of them.

“If you’re part of the project – if you’ve signed up for it and you’re part of the project – provided that information doesn’t affect my children’s ability to get life insurance, mortgages, other aspects of day to day life, then I’ve got no problem with it.”

Male patient, Manchester

Whereas participants were in the position of being relatively established in life, in terms of housing and jobs and so on, it was felt that it was key to young people’s ability to make their way through life to have fair access, without prejudice, to financial services such as mortgages and insurances, and to employment opportunities.

“We don’t know enough about that really...Later on in life, if they said ‘Right, they’ve all had cancer, all the ones in their family, when it comes to the mortgage they’ve got to pay whatever %’. ‘You’re all right, you’ve had no problems, you’re a lesser %’. Why should they have to [pay more] – I mean, they’ve been ill, the chances are they’re not going to earn what you’re going to earn...That’s the only thing that my concern would be.”

Female carer, Manchester

Participants were happy for medical and lifestyle information to be linked with their genome data, understanding intuitively that these details were highly relevant to any medical research. However, the protection of participants’ personal identity was regarded as a basic requirement for involvement in the project, and any prospective decision to take part was seen as contingent on this.
“If you sign the confidentiality clause, you’d like to think that that’s sufficient, wouldn’t you, really. But how do any of us know what information comes out from it or where it goes, really?”

Male carer, Birmingham

Generally, then, participants felt there was a need to be given more information regarding the intended practice of data access and privacy before consent to participate in the project would be given. Although generally participation still was seen as action for the ‘greater good’, there was a wariness of potential catches. It was felt that greater clarity would be needed around exactly how identities would be protected, how data would be accessed and what level of information researchers would be able to view.

“Obviously they don’t need your name, do they – they can have your number, and all the information collated against this number... It is anonymous if that’s the way it’s done, which I imagine it would be.”

Female patient, Birmingham

Perhaps for this reason, participants were not as reassured as they might have been by the analogy, used as part of the stimulus, of the library book that can be read in situ but not removed from the library premises. There was a tendency for participants to read this analogy rather literally, so that they imagined researchers as being a bit like students who would be taking notes as they read, and thus effectively taking away a copy of the protected data. Neither were participants clear that information on volunteers’ personal identities would not be contained in this ‘book’ in any case; further explanation of this is likely to be helpful.

“I don’t think it tells you how they’re going to protect it, other than that they can’t take it away – but you don’t have to take a book away to be able to take the information from it.”

Male carer, Birmingham

The desire was expressed to have a clear separation between personal identifying information and all other data – perhaps even multiple degrees of separation. Some participants began to suggest ways in which the genome data might be tagged with a unique identifying number or barcode, which might in turn be linked to some other anonymised form of marker before tracking through to an individual’s identity.

“I think we’d all prefer it to be anonymous – you know, you don’t want it to go through your name and address... They’re going to have to put in a system...where maybe...they’ve got your hospital number and somehow that’s translated into another reference number – some sort of security process... And then obviously not any researcher can access you – they have to go through Genomics England with a specific code maybe, just for the people doing that research.”
Having said this, there was a pragmatic sense of understanding that every member of society has information ‘out there’ already and that, even with the best will in the world, protection cannot be completely guaranteed.

“No data is totally secure, so I think if you’re going into it you’re going to have to think ‘Well, anything can be hacked.’” If they [hackers] can get into NASA or their systems, then if people want to do it – but we’re probably not as interesting as that – so I think you’re going to have to take that on the chin. But if you can see there is a process in place, that they would be doing things to try to protect you, that’s as much as you can ask really.”

Above all, there was a need for participants to believe that Genomics England would be acting with the volunteers’ best interests at heart, and that everything that can reasonably be done to safeguard confidentiality would be done.

5.3 Feedback and re-contact

There was a wide variation in participants’ views regarding individual feedback, with some participants – both male and female - saying they’d want to know as much as possible about the findings relating to their genome data, whereas others felt they’d prefer to know nothing at all.

“If there was something that could affect you in the future, or possible family members, then you’d want to know... If they found something at gene level, that a normal blood test or something wouldn’t show... If they could say ‘You really need to get these family members looked at’, you’d want to know that information back.”

The drivers behind the desire for individual feedback related to the opportunity to make different choices in life. For example, lifestyle choices and choices about having children, and the opportunity to take pre-emptive action if necessary in order to protect oneself and other family members from illness or disease.

“But wouldn’t you want to know about this HER2? You know, if it came back to - say you haven’t had breast cancer yet, you haven’t got it, but they’ve done this genome thing, they come back and say you’re carrying the gene for HER2... There are these people that find that out and then they have the double mastectomies, don’t they... I would want to know as long as I can do something about it.”

Indeed, a number of anecdotal examples were shared involving participants and people among their acquaintances having been made aware of genetic information that it was possible to act upon. In some cases, this information had been
potentially life-saving. For example, a man in his late 30s had been found, in the course of hospital treatment for an unrelated condition, to have a faulty gene which affected his heartbeat, and for which he was fitted with a defibrillator. Tragically, he had had a brother, now presumed to have had the same gene, who had suddenly died at the age of 14. He also had children of his own, who will be tested as soon as it is practicable.

“But you would [want to know] - because you’d already lost one son, and that [condition] can be fixed, and he’s got two children... That’s a worry, and if the girls have got it they can’t do anything until they’re seven, but if they get through to that age then they can do something for those children... So if you can do something for them... It’s a choice, isn’t it.”

Female patient, Birmingham

It was also hinted, implicitly, that having one’s genome mapped and receiving such feedback information would be a potential side-benefit of participation in the project, analogous to a car having an MOT.

“I think if they’ve [participants have] volunteered to do a trial then they’re obviously curious, aren’t they - so I think that kind of person would want to know anything. I would. It would be like having a full MOT, wouldn’t it.”

Female carer, Manchester

Conversely, the barriers to participants’ wish to receive individual feedback very much centred around a desire to not know about potential conditions.

“I wouldn’t want to know anything, because I’m a born worrier, and if something came back saying to me that, you know, this could happen to your grandchild...I would worry. I wouldn’t want to know. I’d be quite happy to take part, I’d go and have whatever they wanted, but no, don’t want to know.”

Female patient, Birmingham

This was felt to apply particularly if no action could be taken to avert the risk, or where there was a possibility of ‘scaremongering’ - causing worry about risks which might not apply.

“They may be speculating. How do they know that it’s an absolute cast iron guarantee that in three years you might get something? They don’t... It’s a can of worms really.”

Male patient, Manchester

A few participants were of the view that they would be reluctant or unable to cope with additional stress or uncertainty, feeling that they had ‘enough of their plate’ while in the midst of contending with an existing condition.
“I’m not so sure I’d want to know about anything else... Maybe if I wasn’t in the position that I’m in now, I’d probably say ‘Yes, I want to know.’ But I think, going through what we’ve [our family have] all been through these past two years, it’s been bad enough without having to worry about something else, you know.”

Female carer, Manchester

Some participants also felt a desire to protect loved ones from such knowledge, as there might be potential for family members to feel afraid, upset or even ‘guilty’ for passing on faulty genes.

“I don’t think a lot of people would want their family to be scared... If they’re going to pass it [information] on and say ‘Oh, you’re going to have heart trouble through your life’ – you know, you’d be holding the baby going [gestures face falling]... Like I wouldn’t want my parents to get a letter or a phone call to say your family are going to get this or this...because it would destroy them at the moment.”

Male carer, Birmingham

Participants generally felt there was a need to be able to select feedback and re-contact options on an individual basis, in order to reflect each individual’s personal circumstances and preferences. Most participants expected that there would be a form of ‘contract’ setting out such options at the outset.

“But you could also have the question when you sign up – or whoever signed up for this project...yes you do, no you don’t [want feedback]. It would be optional, wouldn’t it.”

Female patient, Birmingham

In addition, it was suggested that in practice such feedback might be wanted by other family members who might be implicated in the findings relating to their relative’s genomic make-up, rather than by the research participant themselves.

“Me, as a carer...I’d want to know - because it’s me and my kids that it’s going to effect - but I wouldn’t want my parents to know, because they’d think ‘I’ve caused this’.”

Male carer, Birmingham

In terms of potential withdrawal options, most participants felt that it would not be fair to impact upon the project as a whole by requiring that existing information and samples be destroyed altogether if a volunteer wishes to withdraw.

“I think that [destruction of data already collected] would be unfair. I think if you sign up for something, you know, somebody’s worked on it for six months or whatever and it’s going to research, to suddenly say – I don’t know why somebody would want to say ‘I want to take it back’: I can’t see that personally. What would it achieve?”
Male carer, Birmingham

However, there was a desire for an option to be available, if requested, to walk away and avoid any further personal contact, and/or to de-link data from personal medical information.

“I’d just think ‘Well, I’ll draw a line under it now. What you’ve got, you’ve got, and I’m just walking away’.”

Female patient, Birmingham

Participants envisaged that the only scenario where they would want to seek the destruction of their records would be in the event of a clear breach of trust, such as an instance of inappropriate disclosure of information.

“You’d want to be able to get out in some way – as much as you would be enthusiastic. You’d want to be able to escape in some way, should something flag up, through the media or whatever, saying that this information’s been used in other ways that you’ve not signed up for.”

Male patient, Manchester

Generally, most participants felt that the obligation be informed of other trials relevant to them was acceptable, as this was not an offer that needed to be taken up.

“I think it [being informed of further trials] would be accepted. It’s only an offer; it’s not set in stone that you have to [do it], is it.”

Female carer, Manchester

However, a couple of participants regarded this condition as being much more problematic. They felt that this obligation created a negative feeling, and said they would prefer not to be ‘pestered.’

“There’s an element there that says to me...once you’ve signed up for this project you’re then bloody fresh meat for any pharmaceutical company that wants to include you in a trial. I don’t like the idea of that at all. It sounds like a time-share! ...I felt ever so slightly more altruistic about it’s probably not a bad idea; this has just [gestures flattening motion] – the shutters have come down.”

Male patient, Manchester

Discussion of the issue of feedback and re-contact was prompted principally in order to gauge participants’ response in relation to information emerging from the mapping of volunteer’s genomic samples. In addition, however, the desire was expressed by some participants to gain feedback on the progress of the project as a whole.
“I think feedback is important, isn’t it. If you’re giving someone information or samples or whatever, it’s important that you get some feedback – you know, not just personally, but how it could help going forward... I can only speak for my partner; there’s an interest in trying to help fight this terrible disease... I think people who have suffered from cancer...I think they’ve got an interest... You don’t expect to get a massive report back but, you know.”

Male carer, Birmingham

The motivation behind this was to feel like a valued ‘member’ of the project community, in much the same way as experienced by donors to cancer charities, and to learn about the successes and milestones towards which volunteers have contributed.

“You’d want to know about successes. You’d want to know that – you’d want information that made you think ‘Yeah, I made a good decision going for that’.”

Male patient, Manchester

5.4 Commercialisation

5.4.1 Commercialisation: Two levels of concern

In the course of discussion of the ethical considerations around commercialisation, two broad themes emerged. The first of these themes focused on the potential for personal impact as a result of participation in the 100,000 Genomes Project, and was very much allied to the issues relating to data access and privacy. The second theme revolved around the profit-making agenda of the commercial companies who would be involved.

The first theme, that of the potential for personal impact, had a much higher level of concern for participants, linked as it was to data access and privacy and the strong desire for anonymity. The fear was that breaches to confidentiality may occur, and that if access to data were gained by third parties such as insurance companies, this could carry the risk of personal detriment and impact on life chances.

“It’s Big Brother, isn’t it... I think a lot of people would go for this, but I also think a lot of those people, I don’t know what %, would say ‘Yeah, you can take my genes, you can take anything you want, but I don’t want to be identified’.”

Female carer, Manchester

The second theme, that of commercialisation in the sense of the involvement in the project of profit-making companies accountable to their shareholders, was a slightly lesser issue for most, though it was a higher concern than perpetuity and non-specific uses.
5.4.2 Commercialisation: Potential for personal impact

The key concerns for participants relating to commercialisation focused upon the potential personal impact it could cause. There was a general assumption amongst participants, however, that the intention would be that information relating to volunteers’ personal identity would remain confidential.

“You’d like to think in the medical world it would be confidential - because it’s amazing how often your name and number and things, companies get hold of them these days. You have to scratch your head and you have to think ‘I wonder how that’s suddenly come, how they got that?’ But you would think that with something like this, that it would be strictly confidential.”

Male carer, Birmingham

Some envisaged genome samples and information being tagged in a coded form as ‘Person A’, with the individual’s medical and lifestyle details linked anonymously to their genome.

But there was a fear amongst participants that they themselves, or their young families, would be vulnerable to discriminatory policies from ‘real world’ companies such as insurers, banks and mortgage lenders, and even employers. This was felt to be potentially detrimental to future life chances.

“I’m sure it’s not going to be ‘Mrs Smith, we know where she lives’ – they’re not interested in that. They just want to know lifestyle and this is the genome... They’re not going to know who that person is. If life insurance companies got hold of it, I’d probably look at it a little bit differently, because they’d be going ‘Ah, you’ve got
three kids - they’ve got a good chance of getting it [cancer]. That I wouldn’t appreciate. But for them trying to make a drug, then...[that’s fine].”

Male carer, Birmingham

An additional fear was that their data would be not just shared but ‘re-shared’ or ‘sold on’ to other third-party companies, who would seek commercial advantage as a result.

“You’ve got too many companies that are selling – obviously your profit-making companies, they’ll sell it to anybody, that information.”

Male carer, Birmingham

This concern appeared to be based largely on people’s personal experiences of direct marketing and being in receipt of overwhelming quantities of junk mail. As an example, one participant had been upset by receiving an appeal to donate to a cancer charity very shortly after his own diagnosis.

“I had a letter through the door, two or three days later, did I want to donate to prostate cancer research? And I thought ‘This is a hell of a coincidence’, you know!... This was at my local BUPA hospital, which you would think would be a tad more controlled than the NHS, I don’t know... And I think that’s probably the danger where this sort of information is gathered: who gets to look at it?”

Male patient, Manchester

Participants were also concerned about data leaks and data being passed to third parties; personal anecdotes were shared concerning the way in which people were able to identify coded markers identifying certain employees as paedophiles, for example. Reference was also made to instances where the controls of government departments over sensitive data have been compromised.

“I think ultimately it doesn’t matter what they do – if someone wants to get it they will get in... It’s only as good as the weakest link... It doesn’t actually change it, because I think, in the world we live in, they’re already doing that to us, whether it’s about genetics or not. The governments or whatever, the supermarkets, they all manipulate you.”

Male carer, Birmingham

As a result, there were questions around how data access and security would be regulated by Genomics England. There was a degree of concern surrounding these ‘weak links’ in data protection, as well as the challenge of imposing data protection controls over multinational and non-UK companies.

“I’m not dead set against – by no means. But would this information only be available to UK-based [companies] or would that be worldwide?... They’re [drugs companies are] all multinational, aren’t they – then you lose control, don’t you,
because data protection’s within this country or Europe, therefore you’ve lost control of your data.”

Male patient, Manchester

5.4.3 Commercialisation: Involvement of profit-making

Although a lesser issue for participants, the involvement of profit-making companies in the project potentially becomes a key concern for some. A degree of scepticism was expressed regarding the high cost of ‘wonder drugs’ and the motivations of commercial companies, who would be driven by the demands of their shareholders.

“I think my only concern with the pharmaceutical companies is, mainly they are looking for making money – you know, the charities are running more for the benefit of those people, so they’re not looking to necessarily make money off the back of it... I don’t know whether I feel the pharmaceutical companies have the best interests at heart really.”

Female patient, Birmingham

This led some participants to question whether these companies should be paying or making contributions to charities and other organisations in exchange for data access.

“If I heard that the drug companies were going to donate money to the charity. I know they’re going to make some profit, but at least if they donated some money to the good cause, then it would be much more interesting.”

Male patient, Manchester

Having said this, the scepticism expressed was set alongside a clear realisation that commercial companies would be in the best position to make actionable developments from the research data, and that they work in partnership with the NHS or government departments already.

“I would hope that this outfit know who they want to be their partners and share this information with. I think I’m probably happy to say that they know better than we do, and they can choose their partners as appropriately as possible.”

Male patient, Manchester

Some participants expressed a pragmatic acceptance of profit-making involvement, given that drug companies have the financial means to develop new treatments. Inevitably, they would profit as a result, but the benefit would be faster medical advancements.

“I think we need to be sort of sanguine about this. We live in a capitalist world – these things are not going to happen unless a big company does them, and a big company isn’t going to do it unless it’s going to make some money out of it. So I
think it’s fine that the government funds a project that produces all the information, but...is the information going to sit on a bloody shelf somewhere unless a big – a Glaxo or whoever – actually do something with it? Yeah? If we’re happy to live in a so-called Western democracy, we have to accept that, you know, it’s about making money... Do we think that it’s a good idea that they save lots of people’s lives? Yeah! Do they make some money whilst they’re doing it? That’s the way the world works.”

Male patient, Manchester

Certainly, however, the involvement in parallel of non-profit making bodies was viewed very positively. Participants perceived that these types of organisations were more likely to have the patients’ best interests at heart, and their involvement was seen as averting the risk of duplication of effort - since charities are also engaged in research, funded by their donors.

“It feels like they’ve [the commercial companies have] almost got to be on board, to be able to push it forward to the depths that they’re going to need to. But it’s nice to know that that [Project] also does get universities and charities involved - because you feel like they’ve [the universities and charities have] got your best interests in mind, the pharmaceutical companies can make it happen. They both kind of need each other.”

Female patient, Birmingham

Again, participants made reference to their concerns at the possibility of third-party access by insurers, banks and other financial companies, employers, media companies and ‘ambulance chasers’ - defined as generating profits from someone’s illness. At the extreme, the depth of feeling surrounding this issue was reflected by a couple of participants expressing a cynical view that Genomics England might be perceived as ‘harvesting’ data and then selling it on to the highest bidder.

5.5 Perpetuity

Access to data in perpetuity was a lesser issue for most participants, who appeared to see assumed ongoing consent as the expected default position for involvement in the project. The commonly held view was that ‘if you’re in, you’re in’, and the data would be retained and used for as long as it was useful.

“If you’re going to go for it, you go for it - or leave it alone. That’s how I feel.”

Male patient, Manchester

There was no perceived requirement for regular reviews of access consent, and most participants were happy to view the granting of ongoing consent as a one-off decision.

“I’m struggling to see why you’d change your mind. If you think it’s a good idea now, what’s going to change your opinion? Apart from Alzheimer’s!”
Indeed, it emerged that several participants had personal experience of having given consent to samples and information being taken for the purposes of medical studies, either in the course of medical treatment, or having been approached in normal daily life, though it was not necessarily something they have thought about since. It is notable that these experiences hadn’t immediately come to mind in the course of earlier discussion about medical research generally, implying an absence of concern subsequent to participation in these previous studies.

“About 3 years ago...I got asked to do a project thing [involving samples of saliva and blood, and measures such as blood pressure, heart rate, weight] and I think I’m to go back in 7 years...I’ve completely forgotten what I signed up to!...But does it bother me? No. I just think ‘I’ve done my bit, there you go’.”

One key proviso was made in relation to this point, however: that if a strong reason to want to withdraw from the project should present itself, there should be provision in place to accommodate this. Some participants referred to extreme scenarios and the impact they would be likely to have on their level of trust – particularly breaches to security and personal privacy.

“I don’t think it matters really – once they’ve got the information, they’ve got it. Whether it’s for a lifetime or not doesn’t really matter, does it, I don’t think...If it came out in the news that they’re selling it [information] to other companies, that’s the only time I could think of retracting it.”

A further example of the type of scenario that it was thought might prompt the wish for withdrawal was that of the stated purposes of the project being altered midway through, so that the research was no longer strictly for the medical purposes being signed up to.

“Well it depends whether they move the goalposts – [if] when you sign up, they said ‘A, B’ and then they went ‘Actually no, we’ve decided it’s going to be C and D now’.”

Some participants also cited personal inability to continue with the project, such as illness or decline in old age, as a reason one might need to withdraw from the project. There were some questions around the level of involvement required, and some felt that ongoing appointments for the purposes of additional data collection, as opposed to one-off donation of the participant’s genome sample, may become a barrier to taking part. This information would need to be given upfront.
“Is it something that they would just do and you just lived your life, or would you have to be involved with it?...If you could just sign something and they just are [working] behind the scenes [that would be fine].”

Female carer, Manchester

Participants envisaged that, as part of an initial contractual agreement to take part in the project, there would be a structured process for withdrawal in place. This would most likely not be used, as they saw it, but would provide a necessary fallback and as such, was regarded as a point of good practice.

5.6 Non-specific uses

There was very little concern expressed regarding non-specific uses of data, provided these were legitimate medical research uses. Indeed, most participants anticipated that there would be an ‘all or nothing’ commitment level to the project.

“There’s no point of view, if I wanted to participate it wouldn’t matter a jot what they did with the data. I would be giving my permission freely.”

Male patient, Manchester

Donation to the project was viewed as taking part in a fight or ‘war’ against disease at a very general level, and for the ‘greater good’ – for medical benefit to anyone, anywhere. Personal benefits were not expected from the project, and there was no sense of an implicit trade-off being made between donation of data and direct benefit to the individual or the individual’s family. Several participants likened this to blood donation; you ‘do your bit’ by giving blood, without knowing who will benefit from your donation.

“I think we’ve already said that it might not necessarily be specific to your condition or to your family. It’s kind of like giving blood, isn’t it – you give blood and you walk away. You don’t want to know where your pint of blood went.”

Male patient, Manchester

Most participants also had a sense that, by definition, the precise outcomes of the project are unknown at this stage. They held the view that the research process, by its very nature, involved ongoing generation and testing of ideas, so that scientists cannot possibly give specifics at the outset on what the eventual outcomes will be.

“I don’t think they could give you specifics on what it would be, because they take the data and then the scientists from then on’ll work out the treatments over years. They couldn’t say, like, your data and everyone else’s data is going to be there, and this’ll be the outcome, because they don’t know the outcome. They get ideas and then they take it further, the scientists.”

Male patient, Manchester
For this reason, non-specific uses were regarded as not only inevitable, but as a positive feature of the project. Most participants viewed it as the means to extract the maximum learning from the data. Furthermore, the potential for advancement in relation to ‘the whole spectrum’ of disease, not just the ‘number 1’ or most ‘headline-grabbing’ conditions, or the ones accorded the most ‘kudos’, was viewed as a distinct benefit.

“I would think that’s the point – you know, for them to look at as much as they possibly can and get as much information out of it as they possibly can... That’s what I’d kind of expect, I think”

Female patient, Birmingham

The view was also expressed that non-specific uses would be necessary to meet the need of researchers to take a more ‘holistic’ view of the data, since it might be that previously unseen links or associations are found to exist between different medical conditions.

“They need to have a holistic view, don’t they, of everything about you and any conditions you may have, because one condition may... have a big effect on another.”

Female patient, Birmingham
6 Concluding points

Although there is some awareness of genetics in relation to crime, genealogy, and hereditary risk factors, there is currently very little awareness of genomics, Genomics England or the 100,000 Genomes Project. Very few were aware of recent headlines, and none mentioned the progression of the Project since the 2003 breakthrough.

The 100,000 Genomes Project was very positively received in concept. Participants felt that proper medical use of data will help fight the battle against disease and ill health. The benefits were not necessarily seen as direct, or personally beneficial to individuals, but instead to be for the ‘greater good’.

Previous experience and understanding of medical research in general represented an important context to participants’ views of the ethical issues. Involvement in the 100,000 Genomes Project was seen as analogous to giving blood. Permissions and consent procedures were therefore less of an issue for sample-based study than for a drug trial, particularly if involvement was to be low level or to take place on a one-off basis.

Participants understood that in order for the 100,000 Genomes Project to contribute to the advancement of treatment, it would be necessary to involve commercial companies and government. People saw this as a trade-off, and although there were some negative connotations, it was generally acceptable: although drug companies will ultimately profit, they will also be responsible for developing new treatments. Central government involvement will help to reduce vested interests and avoid duplication of research.

Participants were keen to be reassured that no personal detriment could come to donors because of their involvement, in the form of personal information being linked to genomic data. There was an understanding that employers, insurers or mortgage providers could potentially use such information to negatively impact on the life chances of donors and their families. Such linking of personal and genomic data was seen as security risk, and privacy breaches were the most likely cause of withdrawal.

There was a spectrum of feeling regarding feedback and re-contact with donors, with some in favour of the idea, and others preferring no follow-up. Most preferred some kind of genome feedback relevant to own and family’s health. There was a desire for clarification and contractual options to be available from the outset of involvement.

Participation in the 100,000 Genomes Project was seen ultimately as an act of good faith. There was a sense that nothing is foolproof, and that rules can only protect you so far. The key appeal of participation lay in the desire to take part for the ‘greater good’ and for the benefit of future generations:

“I think what we’ve discussed more than anything else is that we don’t believe that the rules will actually do anything – so fine, let’s talk about them, but I don’t think any of us actually believe that that’s what really matters. People are going to go in
for this, not because they think the rules are in place. They’ll do it because they’ll think they’re trying to do some good – for the greater good.”

Male patient, Manchester
We are carrying out a Market Research Survey about healthcare and are looking for people to take part in a group discussion. As a token of our appreciation for participation eligible attendees will receive a cash thank you for their time. I just need to ask a few questions first...

**DEMOGRAPHICS**

**Q1a**  Code Gender:

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<td>2</td>
<td>Female</td>
</tr>
</tbody>
</table>

**Groups 1 and 4:**
All to be male at Q1a

**Groups 2 and 3:**
All to be female at Q1a

**Q1b**  How old are you?

*(Write in):*   ___________________________

No Quota (ideally spread in each group) at Q1b

**Q1c**  Are you at present (code all that apply):

Married  1
<table>
<thead>
<tr>
<th>Status</th>
<th>Code</th>
</tr>
</thead>
<tbody>
<tr>
<td>Single</td>
<td>2</td>
</tr>
<tr>
<td>Divorced</td>
<td>3</td>
</tr>
<tr>
<td>Widowed</td>
<td>4</td>
</tr>
<tr>
<td>Separated</td>
<td>5</td>
</tr>
<tr>
<td>Cohabitng</td>
<td>6</td>
</tr>
<tr>
<td>Living with parents</td>
<td>7</td>
</tr>
<tr>
<td>Sharing with friends</td>
<td>8</td>
</tr>
<tr>
<td>Living alone</td>
<td>9</td>
</tr>
</tbody>
</table>

**Q1d**  
Do you have children or are you expecting your first child?  

- Yes, I have children   1    Complete Q1e  
- Yes, expecting first child   2    Continue to Q1f  
- No   3    Continue to Q1f
Q1e Record gender and age of each child living at home:

(Write in): ____________________________________________
____________________________________________________

Record gender and age of each child living away from home:

(Write in): ____________________________________________
____________________________________________________

Q1f Occupation of Chief Income Earner (Probe fully):

(Write in): ____________________________________________
Qualifications: __________________________________________

Number in Charge of: _____________SOCIAL GRADE: ___________

All to be BC1C2 (ideally spread in each group) at Q1f

Q1g If respondent NOT Chief Income Earner ask if:

Working full time (30+ Hours per week) 1
Working part time (6-29 Hours per week) 2
Working less than 6 hours 3
Education (GCSE or pre-GCSE) 4
Education (A-level or equivalent) 5
Education (Vocational - write in): ____________ 6
Education (Degree or equivalent) 7
Education (Post-graduate) 8
Non-Working 9
Retired 10
Other 11
(write in): _______________________

Q1h Occupation of respondent if not Chief Income Earner:

(Write in): ____________________________________________
OCCUPATION/INDUSTRY EXCLUSIONS

Q2 Thinking about the following occupations, can you tell me which, if any:

a) you currently work in or have worked in the past?
   b) any member of your family or close friends currently work in?

Read out: a) b)

Advertising
   X   X
Market Research   X   X
Public Relations   X   X
Journalism   X   X
Marketing   X   X
Healthcare Providers/ Hospitals/
Doctors Surgeries/ Hospices   X   X
Medical Professional/ Doctor/ Nurse   X   X
General/Specialist Medical Practitioner   X   X
Oncologist   X   X
Research/ Manufacture/ Distribution
   of pharmaceutical products or treatments   X   X

__________________________________________

None of the above 0 0

If yes to any responses above the line, close interview
All to code None of the above at Q2a and Q2b

Q2c Do you intend to work in any of those occupations in the next 6 months?

Yes   X   Close
No 2   Continue

PREVIOUS ATTENDANCE

Q3a Are you scheduled to participate in a market research group discussion/depth interview in the near future?
Q3b  Have you ever attended a market research group discussion/depth interview?

Yes  X  Close
No  2  Continue

Q3c  How long ago did you last attend a market research group discussion/depth interview?

In the last 6 months  X  Close
6 Months-3 years ago  2  Ask Q3d
More than 3 years ago  3  Ask Q3d

None to have attended in the last 6 months
Q3d  How many market research group discussions/depth interviews have you attended in total?

_________________

**If more than 3 market research group discussions/depth interviews attended in total close**

Q3e  What was each of those market research group discussions/depth interviews about?

Interviewer write in:

**If on a similar subject as this survey, close interview**

Q4  Can you tell me which of the following medical conditions, if any, of which you:

(a) have yourself had a diagnosis?
(b) care for someone who has had a diagnosis?

<table>
<thead>
<tr>
<th></th>
<th>(a)</th>
<th>(b)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asthma</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Arthritis</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Cancer</td>
<td>3</td>
<td>3</td>
</tr>
<tr>
<td>Diabetes</td>
<td>4</td>
<td>4</td>
</tr>
<tr>
<td>High Blood Pressure</td>
<td>5</td>
<td>5</td>
</tr>
<tr>
<td>Eczema</td>
<td>6</td>
<td>6</td>
</tr>
</tbody>
</table>

**Groups 1 and 3:**
To have had a diagnosis of cancer (code 3) at Q4a

**Groups 2 and 4:**
To care for someone who has had a diagnosis of cancer (code 3) at Q4b
**Ask Groups 1 and 3 only:**

**Q5a** You mentioned you have had a diagnosis of cancer, can you please tell me what kind of cancer?

1. Bladder Cancer
2. Bowel Cancer
3. Brain and Intra-Cranial Cancer
4. Breast Cancer
5. Leukaemia
6. Lymphoma
7. Melanoma
8. Oral Cancer
9. Pancreatic Cancer
10. Prostate Cancer
11. Thyroid Cancer
12. Other (Write In:)___________________

**Group 1:**
Maximum X4 to have been diagnosed with prostate cancer (code 10) at Q5a
PLUS
Good spread of other forms of cancer to be represented at Q5a

**Group 3:**
Maximum X4 to have been diagnosed with breast cancer (code 4) at Q5a
PLUS
Good spread of other forms of cancer to be represented at Q5a

**Ask Groups 2 and 4 only:**

**Q5b** You mentioned you care for someone who has been diagnosed with cancer, can you please tell me what kind of cancer?

1. Bladder Cancer
2. Bowel Cancer
<table>
<thead>
<tr>
<th>Cancer Type</th>
<th>Code</th>
</tr>
</thead>
<tbody>
<tr>
<td>Brain and Intra-Cranial Cancer</td>
<td>3</td>
</tr>
<tr>
<td>Breast Cancer</td>
<td>4</td>
</tr>
<tr>
<td>Leukaemia</td>
<td>5</td>
</tr>
<tr>
<td>Lymphoma</td>
<td>6</td>
</tr>
<tr>
<td>Melanoma</td>
<td>7</td>
</tr>
<tr>
<td>Oral Cancer</td>
<td>8</td>
</tr>
<tr>
<td>Pancreatic Cancer</td>
<td>9</td>
</tr>
<tr>
<td>Prostate Cancer</td>
<td>10</td>
</tr>
<tr>
<td>Thyroid Cancer</td>
<td>11</td>
</tr>
<tr>
<td>Other (Write In:)</td>
<td>12</td>
</tr>
</tbody>
</table>

**Group 2 and 4:**

Maximum X4 to care for someone that has been diagnosed with prostate cancer or breast cancer per group (code 10) at Q5b

**PLUS**

Good spread of other forms of cancer to be represented at Q5b
Ask Groups 2 and 4 only:

Q6    And can you tell who you care for and what kinds of assistance you offer them?

(write in): __________________________________________________________
__________________________________________________________
__________________________________________________________
__________________________________________________________

Record for information purposes only at Q6

Ask All:

Q7    It may be necessary for the research team to contact you by email or telephone after the research has taken place to follow up on ideas generated during the discussion. You would only be contacted if strictly necessary and only in connection with this research. Are you happy to agree to be re-contacted on this basis, and for us to pass your email address to the research team?

Yes                  1  Continue

No                   2  Refer to the office

INVITE TO PARTICIPATE IF RESPONDENT MEETS ALL QUOTAS
RESPONDENT’S DETAILS (PLEASE COMPLETE IN BLOCK CAPITALS)

**Title:** Mr/ Mrs/ Miss/ Ms/ Other: _____________________________________________

**First name:** ______________________________________________________________

**Surname:** ________________________________________________________________

**EMAIL ADDRESS:**  __________________________________________________________

**Mobile Number:**  ____________________________________________________________

**HOUSE NUMBER/NAME:** ____________________________________________________

<table>
<thead>
<tr>
<th>METHOD OF SENDING INVITATION</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>POST</strong></td>
</tr>
<tr>
<td><strong>EMAIL</strong></td>
</tr>
<tr>
<td><strong>FAX:</strong></td>
</tr>
<tr>
<td>(Write in number: ____________ )</td>
</tr>
<tr>
<td><strong>OTHER:</strong></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>CHECKLIST: PLEASE ENSURE YOU HAVE DONE ALL OF THE FOLLOWING BEFORE CLOSING</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Have you?</strong></td>
</tr>
<tr>
<td>(Please circle once completed)</td>
</tr>
<tr>
<td>Re-confirmed time and date of group/depth?</td>
</tr>
<tr>
<td>Given venue details &amp; directions?</td>
</tr>
<tr>
<td>Given Criteria’s telephone number (020 7431 4399)?</td>
</tr>
<tr>
<td>Reconfirmed incentive amount?</td>
</tr>
<tr>
<td>Recorded mobile number &amp; email address? (Where applicable)</td>
</tr>
<tr>
<td>Explained invitation &amp; directions procedure?</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>INTERVIEWER’S DECLARATION</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>INTERVIEWER’S NAME:</strong></td>
</tr>
<tr>
<td>_________________________</td>
</tr>
<tr>
<td><strong>INTERVIEWER’S SIGNATURE:</strong></td>
</tr>
<tr>
<td>_________________________</td>
</tr>
</tbody>
</table>

This interview was conducted in accordance with the Market Research Code of Conduct. The respondent is not a relative or friend of mine.
7.2 Discussion Guide

Genomics England

Discussion Guide FINAL

Research Objectives:

- To explore the views of patients with a diagnosis of cancer and carers on the ethical issues relating to involvement in the 100,000 Genomes Project.
- To specifically explore the following ethical issues:
  - Commercialisation
  - Perpetuity
  - Non-specific uses
  - Data access and privacy
- To identify preferences in terms of data access and feedback.

Note: this discussion guide is intended to inform the discussion with each participant. Questions may not be asked in the order below, and not every question will be asked in each depth interview.

1. **Introductions**

(Aim: introduce participants to the research)

Moderator introduction:

- Thank respondents for taking part in the research, introduce self and GfK NOP.
- Research topic: we are carrying out research on behalf of Genomics England looking at perceptions of ethical issues related to the 100,000 Genomes Project.
- Reassure re: confidentiality, recording and MRS Code of Conduct. We are independent researchers and everything you say will be treated confidentially.
- Explain the group discussion will last 1.5 hours but please let me know if you need to take any breaks or stop for any reason
- Reassure that we are not asking for participation in the 100,000 Genomes Project or trying to convince them to take part.
- Explain importance of being able to say what they think, no right or wrong answers, need for honesty, and validity of opinions.
- If there is anything we discuss that you would prefer not to talk about or any questions you do not feel comfortable answering that is absolutely fine. Please just let me know. You are also free to stop taking part at any time.
- Any questions?

Moderator note:
Adapt the wording of the guide throughout to suit the participants. For example, if the group is with carers, rather than asking about ‘your condition’, ask about ‘the condition of the person you care for’

Also to adapt re-phrasing of questions as appropriate. For example, carers should be asked how they might advise the person they are caring for rather than asking about their involvement.

2. **Participant Introductions**  
(Aim: get to know more about the participants’ life and their condition/person they care for)

It would be great to start off by getting to know each other a little better…

- First name, age
- Family
  - Who they live with
  - Children age(s)
  - Pets
- Work and career, home life (home duties)
- How do they spend their leisure time and who with?
  - Hobbies/ things enjoy doing
- Condition
  - Treatment receiving/received
  - How you/the person you are caring for is at the moment

3. **Initial attitudes towards medical projects**  
(Aim: to gauge initial perceptions and attitudes towards medical testing/trials)

I would like to start by asking your opinions towards medical studies/drug trials on humans…

- What have you heard about medical studies/projects/trials?
  - Where have you heard about it?
  - What would be the reasons for/against taking part in it?
  - Would you/the person you care for ever take part in one?
- Have you/the person you care for ever been involved in a medical project/study/drug trial before?
  - If no: is there a reason why you have not taken part?
  - If yes: could you tell me a little bit about it?
    - Who was running the project/study/trial?
    - What was it for?
    - How long was it for?
    - Why did you take part?
- Do you think medical studies/projects/trials are of benefit?
  - Why do you say that?
  - Who would they/wouldn’t they be of benefit?
• Do you have any concerns about medical studies/projects/trials?
  o Probe: safety, ethics, profit-making, access
  o Why do you have these concerns?

**Exercise 2: Barometer exercise**

I am now going to hand out an exercise I would like you to quickly fill out. Based on what we have discussed, please could you mark on the barometer how you feel about your participation in medical trials/projects/studies on the barometer? The higher you put your mark on the barometer, the more positive you feel about what we have discussed. If you could write why you have marked that particular spot would be great too.

4. **Genomics England and the 100,000 Genomes Project** 10 mins
(Aim: to ensure similar levels of understanding amongst participants. To allow for initial concerns/questions to be raised.)

I would now like to show you some information about Genomics England and what they do.

**Showcard 1-3:**

- Had you heard about this information before?
- Had you heard of the Human Genome Project before?
  - Where did you hear about it?
  - What did you hear about it?
- How do you feel about the study of genomes and genetics?
  - Is this something you feel is important?
  - Why/why not?

**Showcard 4-5:**

- Had you heard about this information before?
- How do you feel about linking a genome sample to other personal information?
  - Would you have any concerns about this? Why/why not?
  - What would those concerns be?
  - Do you feel that it is needed to link up to other information?
  - Would any measures put in place reassure you about the usage of personal information?
    - What would these measures be?
    - Why would they reassure you?
- How do you feel about this type of medical research being linked to cancer?
  - Why do you say that?

**Showcard 6-7:**
• Had you heard of Genomics England before?
  o If yes: where did you hear about it?
  o What had you heard about it?
• Had you heard of the 100,000 Genomes Project before?
  o If yes: where did you hear about it?
  o What did you hear about it?
• What do you think of Genomics England and their work after reading that?
  o How do you feel about Genomics England being set up by the Department of Health?
  o What do you think about the work they are trying to achieve?
  o What do you think about Genomics England working with those who have had a diagnosis of cancer?
  o What are your thoughts on the benefits of the study?
    ▪ Would you/the person you care for take part in a study where you may not benefit personally?

• Do you have any questions or concerns about the information you have just read?
  o Why/why not?
  o What other information would you like to know?
• Would you/the person you care for take part in the 100,000 Genomes Project?
  o Why/why not?
  o What else would you need to know?

**Exercise 3: Barometer exercise**

I would now like you to fill out the barometer exercise based on the information I have just shown you. Based on what we have discussed, please could you mark on the barometer how you feel about your participation in the 100,000 Genomes Project? The higher you put your mark on the barometer, the more positive you feel about what we have discussed. If you could write why you have marked that particular spot would be great too.

5. **Ethical Issues** 40 mins

(Aim: to understand perceptions and attitudes of ethical issues related to the 100,000 Genomes Project and whether this would affect participation)

I would now like to discuss with you some ethical questions or concerns that are related to the 100,000 Genomes Project. I am going to read out some information and it would be great to get your views and how Genomics England could take steps to reassure you or address these questions/concerns.

*Moderator note:*
COMMERCIALISATION

Moderator to read out:

Imagine that you are/the person you care for is considering taking part in the 100,000 Genomes Project. The researchers who are running this project would allow commercial (profit making) companies, such as pharmaceutical companies, access to information about the genes and health of participants with cancer. This is because these companies are responsible for developing the vast majority of cancer drugs and diagnostic tests in use in the NHS today.

Researchers would also come from universities, charities or other organisations who are funded either by the taxpayer, research funding charities, or by donations from the public. These universities, charities or other organisations are unlikely to have stakeholders or directly seek to make money from their research.

- How would you/the person you care for feel about your health information being shared with commercial/private companies?
  - Do you see any benefit for your health information being shared with these types of companies?
- Which companies or organisations do you/the person you care for feel should be allowed to access this type of health information?
  - Why do you say that?
  - What is the benefit for these types of companies or organisations to access this type of information?
- Which companies or organisations do you/the person you care for feel should not be allowed to access this type of health information?
  - Why do you say that?
  - Would you feel differently about the project if these companies/organisations were not allowed to access this information?
    - Why do you say that?
    - Would you/the person you care for consider taking part in the project?
- If you/the person you cared for volunteered to take part in this project, do you think the sharing of your information with third parties/commercial companies should be optional?
  - Why do you say that?
  - If the decision was optional, would you feel differently about taking part in the project?

Exercise 4: Barometer exercise

I would now like you to fill out the barometer exercise based on what we have just discussed. The higher you put your mark on the barometer, the more positive you feel about what we have discussed. If you could write why you have marked that
particular spot would be great too.

<table>
<thead>
<tr>
<th>Question</th>
<th>Response</th>
</tr>
</thead>
<tbody>
<tr>
<td>Does this change how you feel about the 100,000 Genomes Project?</td>
<td></td>
</tr>
<tr>
<td>Does this change how you feel about Genomics England?</td>
<td></td>
</tr>
<tr>
<td>How likely would you/the person you care for participate in this project based on this information?</td>
<td></td>
</tr>
<tr>
<td>What would genomics England have to do to reassure you?</td>
<td></td>
</tr>
</tbody>
</table>

**PERPETUITY**

**Moderator to read out:**

The 100,000 Genomes Project would ask participants to allow researchers to have access to their NHS and other records and do the information from their DNA samples, for the whole of their lifetime and after their death.

- How would you/the person you care for feel about giving researchers permission to access your information from the NHS and other records, and to a sample of your DNA, for your whole lifetime and after your death?
  - How do you feel about this time period?
  - What do you think would happen if you decided in the future you no longer wanted researchers to access your samples and information?
  - Would you suggest a different arrangement?
    - For example, you give consent to access information for five years at a time.
- Is there a situation in the future where you/the person you care for might decide you no longer want researchers to be able to access your health information for your whole life and after your death?
  - For example: would you feel differently about continuing with the project if analysis of your genome did not indicate a specific treatment option for you?
  - What might this situation be?
  - How do you think you would go about telling Genomics England you no longer want researchers to access your information?
  - How do you think Genomics England would respond to your request?

**Exercise 5: Barometer exercise**

I would now like you to fill out the barometer exercise based on what we have just discussed. The higher you put your mark on the barometer, the more positive you feel about what we have discussed. If you could write why you have marked that particular spot would be great too.

<table>
<thead>
<tr>
<th>Question</th>
<th>Response</th>
</tr>
</thead>
<tbody>
<tr>
<td>Does this change how you feel about the 100,000 Genomes Project?</td>
<td></td>
</tr>
<tr>
<td>Does this change how you feel about Genomics England?</td>
<td></td>
</tr>
<tr>
<td>How likely would you/the person you care for participate in this project based on this information?</td>
<td></td>
</tr>
<tr>
<td>What would genomics England have to do to reassure you?</td>
<td></td>
</tr>
</tbody>
</table>
NON-SPECIFIC USES

Moderator to read out:

The 100,000 Genomes Project would give researchers access to participants’ information for medical research into their specific condition. Researchers would also be granted access to this information in order to do medical research in general. For example, in to conditions that may not be relevant to you or your family.

- What do you think about this?
- How important would it be for the research to be of benefit specifically for you/the person you are caring for and your family?
  - What if the research had no specific benefit to you and your family?
    - Probe: may advance scientific understanding (e.g. finding a better treatment) in to your condition, but you/your family may not directly benefit
  - Would you only consider taking part in this project if the research had a direct benefit to you/your family?
  - How would you feel about researchers accessing your information?
    - Probe: does this feel like a ‘trade off’ in order to find a better treatment for you/your family?
- How do you feel about your data being used to research other conditions?
  - What difference does this make to you/the person you care for?
  - How would you feel about researchers accessing your information to use for research into any medical condition?

**Exercise 6: Barometer exercise**

I would now like you to fill out the barometer exercise based on what we have just discussed. The higher you put your mark on the barometer, the more positive you feel about what we have discussed. If you could write why you have marked that particular spot would be great too.

- Does this change how you feel about the 100,000 Genomes Project?
- Does this change how you feel about Genomics England?
- How likely would you/the person you care for participate in this project based on this information?
- What would genomics England have to do to reassure you?

DATA ACCESS & PRIVACY

Moderator to read out:

In order to access Genomics England’s data services, both academic and commercial researchers will have to apply to a data access group. Genomics England would check thoroughly that the researchers’ application is for a legitimate medical purpose. When researchers access Genomics England’s data services, they
would not be allowed to take anything away from the Genomics England’s facilities. For example, think of a book that you are not allowed to take out of a library, but you are allowed to look at it in the library.

- Overall, how important is the privacy of your medical information to you/the person you care for?
  - What arrangements/procedures would you like to see in place in order to safeguard your information?
  - Are there any companies or organisations that you/the person you care for would not wish to access your data?
    - Probe: even if Genomics England carried out the arrangements and procedures I described to you?
- How do you/the person you care for feel about researchers applying to access the data and Genomics England checking their application?
  - Is this something that is important to you?
  - Is this measure satisfactory?
  - How reassuring is this?
  - Would you/the person you care for trust Genomics England to check the application to a standard you would be happy with?
  - Would you/the person you care for trust Genomics England to only allow researchers access to the data for legitimate medical purposes?
  - What else could Genomics England do?
- How do you/the person you care for feel about Genomics England not allowing researchers to ‘take’ any data away from the Genomics England facilities?
  - Is this something that is important to you?
  - How reassuring is this?
  - Is this measure satisfactory?
  - Would you trust Genomics England to ensure that data is not taken off of the premises?
  - What else could Genomics England do?

Moderator to read out:

Genomics England will do everything it can to make sure that your medical information cannot be identified as belonging to you. However, if someone has a particularly unusual or rare condition, it might be possible to link the medical information to an individual, and therefore Genomics England cannot absolutely guarantee data privacy. However, it is illegal to reveal the identity of someone this way and Genomics England would impose its own sanctions if a researcher did this.

- How important is it to you/to the person you care for that your identity is kept anonymous?
- How reassuring is this?
- Is this measure satisfactory?
- Do you feel that Genomics England would do everything it could to protect your identity from being revealed?
• How do you feel that someone’s identity may be revealed if they have an unusual or rare condition?
• Do you feel that Genomics England would impose its own sanctions if a researcher did reveal an individual’s identity?
• Is there anything else that Genomics England could do to better protect the identity of those who took part in the project?

**Exercise 7: Barometer exercise**

I would now like you to fill out the barometer exercise based on what we have just discussed. The higher you put your mark on the barometer, the more positive you feel about what we have discussed. If you could write why you have marked that particular spot would be great too.

- Does this change how you feel about the 100,000 Genomes Project?
- Does this change how you feel about Genomics England?
- How likely would you participate in this project based in this information?
- What would genomics England have to do to reassure you?

**FEEDBACK & RECONTACT**

Moderator to read out:

Imagine you are/the person you are caring for is considering taking part in the 100,000 Genomes Project.

- What information would you/the person you are caring for like to receive:
  - Only information relevant to your/their cancer?
  - Any aspects of your/their genome relating to cancer that might have relevance for the health of your family/future family
  - Any aspects of your/their genome that might have relevance for the health of your family/future family
  - Other aspects of your/their health unconnected with cancer
    - Anything relevant
    - Only things where some action could be taken to help

Moderator to read out:

Imagine you have/the person you are caring for has joined the 100,000 Genomes Project but have since changed your/their mind and you want to withdraw from the project.

- Would you/they like to have the choice of:
  - No further use: destroy all samples and delete information held about me
  - No further access: unlink my samples to my health records but researchers can still have access to my samples and to previously collected information
  - Don’t contact me again: access to my samples and health records but I do not want any contact
I feel that the only options should be consent to full participation or no further use and destroy all samples and delete information held about me.

• Are there any other choices that you think people should be given or may like to choose?
  o What are they?
  o Why do you think people would like to have the choice you suggested?
• What scenarios/situations do you think may happen in the future which may make people change their minds about participation in the project?

Moderator to read out:

The 100,000 Genomes Project will require that participants agree to be told by their treating clinician of further studies or drug trials that they are eligible for. There is no obligation for you to take part in any of these.

• How would you/the person you are caring for feel about your treating clinician telling you of further studies/drug trials that you would be eligible for?
• How do you/the person you are caring for feel about this requirement being mandatory?
• Would this change your/their attitude towards participation in the project?
  o Why/why not?

Exercise 8: Barometer exercise

I would now like you to fill out the barometer exercise based on what we have just discussed. The higher you put your mark on the barometer, the more positive you feel about what we have discussed. If you could write why you have marked that particular spot would be great too.

• Does this change how you feel about the 100,000 Genomes Project?
• Does this change how you feel about Genomics England?
• How likely would you participate in this project based in this information?
• What would genomics England have to do to reassure you?

6. Attitudes towards medical projects and Genomics England  10 mins
(Aim: to gauge whether perceptions and attitudes towards the project have changed throughout)

I would like us to discuss your opinions about medical studies/drug trials on humans again, now that we have spoken about the 100,000 Genomes Project...

• Have your opinions changed about medical project/study/drug trials?
  o If no: why not?
  o If yes: how have your opinions changed?
    ▪ Has your opinion changed positively/ negatively?
    ▪ Did you learn something today that you did not know before?
• Do you think medical studies/projects/trials are of benefit?
  o Why do you say that?
  o Who would they benefit?
Do you have any concerns about medical studies/projects/trials?
- Probe: safety, ethics, profit-making, access
- Why do you have these concerns?
- Has this discussion highlighted any concerns that you did not previously have?
- Has this discussion answered any concerns that you previously had?

**Exercise 9: Decision maker**

You are the main decision maker at Genomics England and you can make the rules about accessing individual’s information and the feedback they receive. In an ideal world:

- What would the rules be:
  - Who is allowed to access the data and information
  - When and how they can access it
  - The length of time your data and information is stored and accessed for
  - How your data and information is used (whether it is for specific conditions or for all medical purposes)
  - Anonymity and privacy
  - The feedback an individual receives about their genome
  - Being told about other medical trials/studies that an individual could participate in
  - Withdrawing from the study in the future
- Why have you decided these rules?

7. **Wrap up** 5 mins

(Aim: gather any other comments/ wrap up the discussion)

Gather final thoughts and comments.

**Thank and Close**

**Signposting:**

For those who would like more information about Genomics England.

Website: [www.genomicsengland.co.uk](http://www.genomicsengland.co.uk)

Telephone: 0207 882 6392

Email: [info@genomicsengland.co.uk](mailto:info@genomicsengland.co.uk)
7.3 Show cards

What is Genetics?

Genetics is the study of the way particular features or diseases are inherited through genes and passed down from one generation to the next. Genes usually work together as groups and their activity is influenced by a huge variety of factors, including environmental.

The Human Genome Project

In April 2003 it was announced that a complete map of the DNA of a person – their genome – had finally been finished. This work was known as The Human Genome Project.

This scientific discovery about DNA and the way it works could revolutionise medical treatment in the future.

What is Genomics?

You have a complete set of genes in almost every cell in your body. One set of all these genes is called a genome. Genomics is the study of the whole genome and how it works.

Studying Genomics

The Human Genome Project demonstrated that before the project, 95% of a person’s genome was a mystery and it is very important to sequence the whole human genome to understand the role of genes in health and disease.

But people are different, and studying genomes alone cannot tell doctors and scientists very much about our genes and their relationship to disease. To make sense of it, it is essential to know more about the person who donated the genome. Information that would be very useful in understanding a person’s genome include:

- Physiological measurements
- Details of symptoms and when they first started
- Information such as blood pressure
- Any other useful information such as previous illnesses, medications and other medical records.
What can genomics do?

Genomics can be used to predict how well a person will respond to a treatment or find one that will work best for them.

An existing example of this is whether or not a woman’s breast cancer is HER2 positive. If it is, Herceptin will be very effective for her but not for someone who doesn’t have HER2.

Genomics can also be used to track infectious disease, and precisely pinpoint the source and nature of the outbreak.

The potential of genomics is huge, leading to more precise diagnostics for earlier diagnosis, new medical devices, faster clinical trials, new drugs and treatments and potentially, new cures.

Who is Genomics England?

Genomics England was set up by the Department of Health to deliver the 100,000 Genomes Project.

By 2017, the project will sequence 100,000 genomes from patients with rare disease or cancer. For cancer, it will mean greater insight into its cause, progression and most effective treatment.

Some participants with cancer may benefit personally but it is more likely that participants will benefit people like them in the near future through new medicines, treatments and diagnostics.

Undertaking this very large project will also make it possible for the NHS to have a genomic medicine service by 2018, so that any sick person who needs it will be able to use it in the future.

Why are cancer patients included in the 100,000 Genomes Project?

Cancer begins because of changes in genes within what was a normal cell and develops mutations or changes.

DNA can be taken from the tumour and from the person’s normal cells in order to be compared to look for changes. Knowing and understanding these changes strongly indicates which treatments will be the most effective.

Genomics has already started to guide and inform doctors about the best treatment for individual patients.
BAROMETER 1

| Your name ________________________________ |

Barometer exercise ____

On a scale of 1 – 10, how positive do you feel about what we have discussed? 1 is not at all positive and 10 is very positive. Please circle a number.

1 2 3 4 5 6 7 8 9 10

Why have you marked that particular number? __________________________________________

_________________________________________________________________________________

_________________________________________________________________________________