

# Project Report

NHS Genomic Medicine Centres  
National Service Evaluation of the Consent  
Process and Participant Materials used in  
the 100,000 Genomes Project

**NHS**  
*North West Coast*  
*NHS Genomic Medicine Centre*

## Authors

Caroline Benjamin<sup>1</sup>, Markella Boudioni<sup>1</sup>, Helen Ward<sup>1</sup>, Elliot Marston<sup>1</sup>, Antje Lindenmeyer<sup>1</sup>, Munirah Bangee<sup>1</sup>, Julie Cook Lucas<sup>1</sup>, Ralph Leavey<sup>1</sup>, Mark Caulfield<sup>2</sup>, Tom Fowler<sup>2</sup>, Anneke Lucassen<sup>2</sup>, Fiona Rennie<sup>2</sup>, Laura Riley<sup>2</sup>, Michael Parker<sup>2</sup>, Vivienne Parry<sup>2</sup>, Ellen Thomas<sup>2</sup>, Christine Patch<sup>2</sup>, Alison Cranage<sup>3</sup>, Lisa Dinh<sup>3</sup>

<sup>1</sup> Service Evaluation Project Team

<sup>2</sup> Service Evaluation Steering Group

<sup>3</sup> Genomics England Support for the Evaluation

**November 2016**

## Contents

<b>Foreword from Professor Michael Parker</b>	<b>3</b>
<b>Foreword from Professor Sue Hill</b>	<b>4</b>
<b>Evaluation Summary</b>	<b>5</b>
<b>1 Background</b>	<b>12</b>
1.1 Context of Consent in the 100,000 Genomes Project	12
1.2 Context of the National Service Evaluation Project	12
1.3 Shared purpose and scope of project	13
<b>2 Methods</b>	<b>14</b>
2.1 Evaluation Methodology	14
2.2 Focus Group Methods	17
2.3 Health Professional Survey Methods	17
2.4 Participant Feedback Methods	18
<b>3 Results</b>	<b>22</b>
3.1 PPI Focus Groups	22
Topic area 1 - Logical flow of consent documentation	22
Topic area 2 - Complexity of project structure	24
Topic area 3 - Seeking broad consent for additional findings	26
Topic area 4 - General transformational and sustainability issues	27
Additional barriers and facilitators	29
3.2 Health Professional Recruiter Survey	31
Summary	37
3.3 Participant Survey	38
Rare Disease & Cancer (n = 172)	39
Summary	41
<b>4 Synthesis of Activities</b>	<b>42</b>
Synthesis methods	42
Visual Mapping	44
Synthesis Tables	45
Synthesis summary	50
<b>5 Recommendations</b>	<b>51</b>
<b>Acknowledgements</b>	<b>54</b>
<b>Glossary</b>	<b>55</b>
<b>References</b>	<b>55</b>

## Foreword from Professor Michael Parker

In December 2012, I was asked by the Chief Medical Officer for England, Professor Dame Sally Davies, to chair an Ethics Advisory Group on Genomics leading up to the establishment of the 100,000 Genomes Project. We were given the task of identifying the key ethical challenges that would need to be addressed as part of ensuring the successful and ethical completion of the Project. We were also invited to make recommendations about how these challenges might be resolved. It was clear from the outset that the 100,000 Genomes Project was going to need to be innovative, not only in its application of genomics at scale in the clinical setting and in research, but also in its development of an approach to good ethical practice in genomics.



One of the most important recommendations of the Advisory Group was that the successful completion of the 100,000 Genomes Project and the sustainable introduction of genomics into the NHS were going to depend upon the development of an effective model for the achievement of valid consent in genomics at the clinical-research interface. We were aware at that time that whatever model of consent was adopted by the project would need to be complemented by regular evaluation and would be likely to need to be modified over time informed by good quality evidence about its effectiveness and acceptability to patient-participants.

Against this background, I am absolutely delighted to have been asked to write a foreword to this important report, which presents the findings of a study led by three of the Genomic Medicine Centres on the national experiences of three stakeholder groups: revealing the first evidence we have of these for patient-participants in the 100,000 Genomes Project, recruiters, and Patient and Public Involvement groups in the NHS.

This is a vitally important and timely piece of research as we review the participant materials in the light of the emerging available evidence. We were reassured to see that (for example) the majority of responding participants endorsed the length and content of the information materials. The majority of responding recruiters similarly endorsed the content, but prioritised brevity of these materials, which we will note for future iterations.

We warmly welcome this contribution to the evidence base for consent in genomics in clinical and research practice.

A handwritten signature in black ink that reads "M Parker". The signature is fluid and cursive, with a large, sweeping initial 'M'.

Professor Michael Parker  
Professor of Bioethics and  
Director, The Ethox Centre  
University of Oxford

## Foreword from Professor Sue Hill



The 100,000 Genomes Project is at the heart of our efforts to transform healthcare in England from a one-size-fits-all system to a targeted, personalised approach where patients will get individualised treatment based on their unique characteristics. NHS England has established 13 NHS Genomic Medicine Centres to cover the country delivering an end-to-end genomic medicine pathway and they are enabling the participation of patients and family members in the 100,000 Genomes Project with their informed consent; collecting samples to extract DNA; capturing clinical information to inform the interpretation of the genome sequence; and establishing the infrastructure to make genomic medicine a routine part of NHS care.

The information consenting process itself is key to the patient experience and to supporting healthcare professionals. This has been identified consistently as an area in which there is scope for improvement if we are to transform ways of working and build whole genome sequencing as part of the genomic testing repertoire into routine patient pathways.

I am delighted that three of the NHS GMCs have led this piece of work to evaluate the consent materials.

I would like to say thank you to everyone who contributed to the evaluation, recognising that the insight gained from recruiting professionals, participants and patient and public involvement (PPI) groups has provided a more robust evidence base from which to revise the materials.

We are very pleased to note that for example, 87% of staff who completed this survey felt 'confident' or 'very confident' in being able to provide a quality consent experience. We will be working with colleagues at Genomics England to take on recommendations such as addressing the literacy needs and providing easy to read versions of the materials.

We have also recently committed to resourcing a national 100,000 Genomes Project Information Line, operated initially by South West NHS GMC. This directly supports the recommendation in the report to explore ways of ensuring that recruiting staff have appropriate time to discuss the issues raised in the Participant Information Sheet (PIS) with participants, where this is needed.

This project is hugely important to the international community who are undertaking similar projects and considering the introduction of Whole Genome Sequencing into routine clinical care. The appropriateness of informed consent of patients and listening to participants is vital to implementation and success.

A handwritten signature in black ink that reads "Sue Hill". The signature is written in a cursive style with a long, sweeping underline.

Professor Sue Hill OBE  
Chief Scientific Officer  
NHS England

## Evaluation Summary

This report details the outcome of a snap-shot national service evaluation which aimed to gather feedback and determine the facilitators and barriers related to the perceptions and processes of those involved in consent conversations during the initial stages of recruitment into the 100,000 Genomes Project. Data collection was undertaken between May and July 2016.

In 2013, The Prime Minister launched the 100,000 Genomes Project to bring the predicted benefits of genomics to NHS patients. Genomics England, a company owned and funded by the Department of Health, was set up to deliver this flagship project which will sequence 100,000 whole genomes from NHS patients by 2017. Its four main aims are:

- To create an ethical and transparent programme based on consent;
- To bring benefit to patients and set up a genomic medicine service for the NHS;
- To enable new scientific discovery and medical insights; and
- To kick start the development of a UK genomics industry.

The 100,000 Genomes Project focuses on patients with a rare disease and their families, and patients with cancer. The project was the first to be processed through the Health Research Authority as a National Health Service Transformational Project. As such it has a hybrid status; that of requiring research governance but also transforming clinical practice.

The first samples for sequencing were taken from patients living in England in April 2015. Recruitment for rare disease patients had been ongoing for 12 months, and 2 months for cancer patients prior to this evaluation.

A commitment to evaluate the consent materials was outlined in section 12.2.1 of the 100,000 Genomes Project approved Protocol v 2.0, dated February 2015: 'Genomics England intends to further evaluate and revise its literature, and patient and public materials at the 10,000 participant recruitment point' (Available from <https://www.genomicsengland.co.uk/library-and-resources/>). The development and methodology of this National Consent Evaluation is detailed in a separate document, the 'Service Evaluation Schedule', available from [www.genomicsengland.co.uk/consent-evaluation](http://www.genomicsengland.co.uk/consent-evaluation).

Three stakeholder groups were involved from across 13 NHS Genomic Medicine Centres (Figures 1 and 2). This social and behavioural science research follows a mixed methods design with parallel mixed methods data analysis culminating in the development of meta-inferences (conclusions generated through an integration of the inferences that were obtained in the three strands of the study).

Figure 1 Three stakeholder groups were involved: patient and public involvement groups associated with Genomic Medicine Centres, Health Professional recruiters and 100,000 Genomes Project participants themselves.

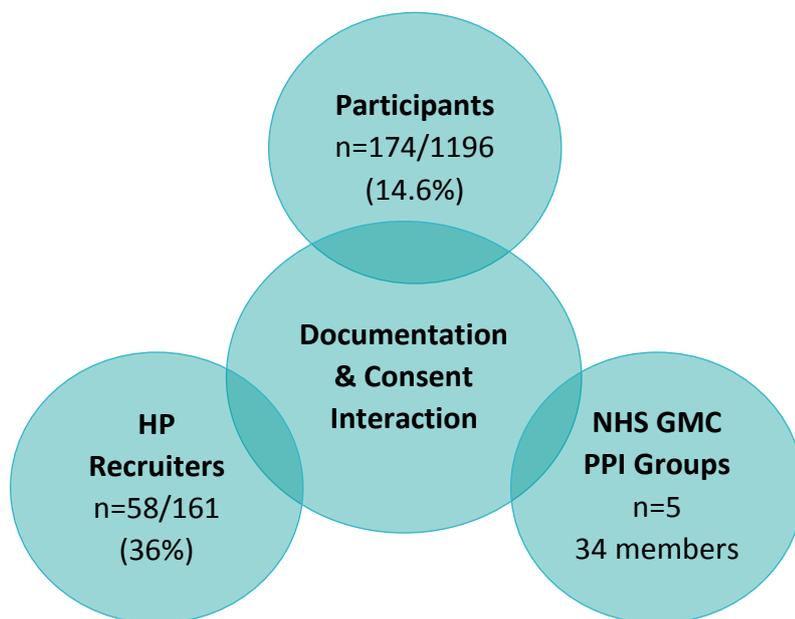
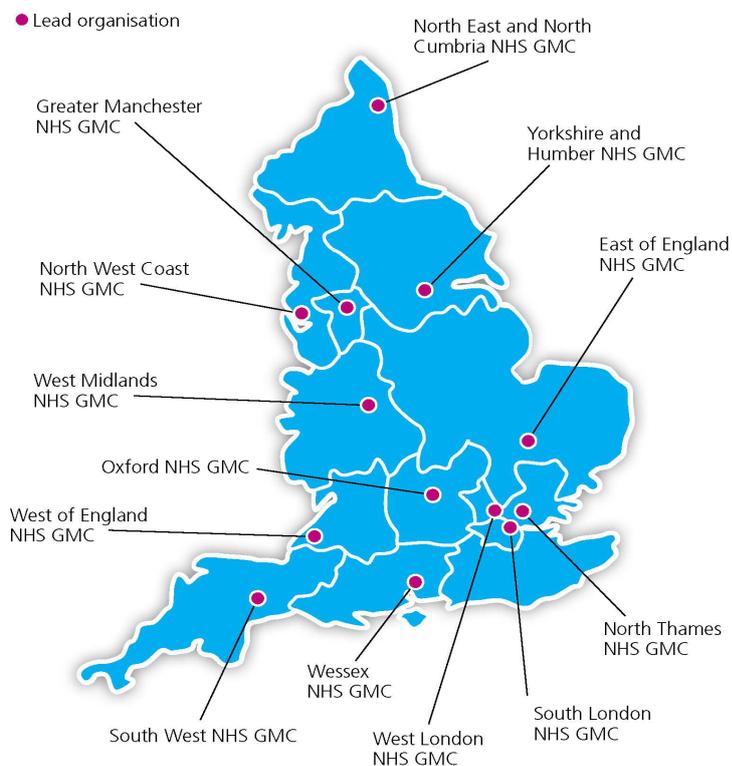


Figure 2 Thirteen Participating Genomic Medicine Centres

## NHS Genomic Medicine Centres



Paving the way to personalised medicine



## General

- Good communication was established with the Genomic Medicine Centres and all engaged in some way with this 8 week snap-shot evaluation project.
- The evaluation succeeded in eliciting views from three stakeholder groups involved in the consent process. Quantitative survey results should be viewed with caution due to low response rate and possible sources of bias.
- On occasion views diverged between and within stakeholder groups, however there were also similarities.
- The consent process varied as to how it was implemented between different GMCs and there was evidence of local process adaption.

This snap-shot evaluation (mid-way through the 100,000 Genomes Project) showed that there were differing opinions both within and between stakeholder groups. This means that when considering the development of materials to help facilitate consent, it is important to bear in mind that how they are perceived and utilised will differ in relation to context and the individual participant's information needs. The results need to be considered alongside on-going and published research as well as ongoing policy debate regarding consent for whole genome sequencing.

There was support for the supplementary materials developed to help with the consent process, such as the information leaflet, animations and flip-chart. These were viewed as being useful in preparing patients for the consent discussion. Suggestions were provided concerning how to support NHS recruiting staff, including on-going training. Overall, 87% of staff who completed this survey (who represented 36% of active recruiting staff at the time of the evaluation) felt 'confident' or 'very confident' in being able to provide a quality consent experience. Although not necessarily representative of all participants, due to the response rate of 14.6%, those 100,000 Genome Project participants who returned a feedback postcard (n = 172) were extremely satisfied with the consent materials and process they experienced. Participants in the focus groups were also supportive of the information included in the consent material. There were aspects of the consent materials and consent process that received positive comment from all stakeholder groups.

However, evidence of where the materials could be improved was also found. Concern was raised by recruiters and focus group members regarding the length and readability of the participant information sheets (PIS) and consent forms, and they proposed specific changes that could be made. These are detailed in the appendices of the 'National Service Evaluation Project: Background Document' which is available to Genomics England for review when implementing changes to the consent material or process.

Half or more of the recruiters perceived that participants 'sometimes', 'often' or 'very often' experienced difficulty with certain sections of the consent forms, especially the sections 'taking part', 'samples', 'access to my data and confidentiality', and 'results'. Approximately half of the recruiters reported that participants 'often' or 'very often' needed 'extra explanation to understand the project', had specific needs related to 'low levels of literacy' and 'increased levels of anxiety related to current health situation'. All stakeholder groups felt it was important to keep most of the current informational content in some way within the materials but wished for changes to the PIS and consent documents in terms of presentation and readability.

Figure 3 provides an infographic overview of some of the evaluation findings.

**The following recommendations are made to Genomics England and are based on the evidence from this evaluation. It is expected that Genomics England will take forward the conversations with partners across the healthcare system such as NHS England, Genomic Medicine Centres and Health Education England.**

Recommendations 1 to 8 are suggestions on elements to maintain, recommendations 9 to 17 are issues Genomics England may consider changing in relation to project documentation, and recommendations 18 to 26 are linked to project processes. Each recommendation is linked back to the data collected by providing the recommendation number alongside the evidence within the synthesis tables in Chapter 4.

#### **Things to maintain (1-8)**

1. The current range of consent materials i.e. participant information sheet (PIS), consent forms, information leaflet, animation/flip chart.
2. The current wording in the PIS relating to explanations of scientific and genomic terminology in line with recommendation 9.
3. The majority of the information content within the PIS (responses from participants and most focus group members support this, along with 50% of recruiters).
4. The PIS both in paper form and on-line (not just on-line).
5. The key point boxes and colour on the forms.
6. Continue to develop accessibility to consent materials for low vision patients.
7. The ability of participants to opt-in and opt-out of the search for additional findings.
8. The facility for health professionals to have the time to adequately discuss and answer patient questions prior to the consent decision.

#### **Changes to consider - Project Documentation/Accessibility (9-16)**

9. Improve the presentation and readability of the PIS and consent forms in the following ways.
  - 9.1 Reduce the density of words.
  - 9.2 Reduce replication of text especially the sections between the consent form and PIS and also after the key point boxes.
  - 9.3 Use pictures/diagrams where possible.
  - 9.4 Work with participants/100,000 Genomes Project National Participation Panel/PPI groups/Ethics Advisory Committee to review new material/changes to assist in approaching standards such as the Plain English Guidelines and the Accessible Information Act.
  - 9.5 Reduce the length of the consent form.
  - 9.6 Review the terminology (e.g. main findings, additional findings) used across all project materials, including written and animation/video to assist with understanding the project specific terms.
  - 9.7 Include a source for standard definitions/concepts (this could also be available for recruiters) for example, a glossary of definitions of both scientific and project terminology and ensure these are consistent with descriptions in the text.
10. Continue to support non-English speakers by promoting the use of existing NHS translation services when discussing project documentation with potential participants.
11. Produce an 'Easy Read' version of the PIS and consent form that could be used for participants with reduced intellectual ability or those for whom English is a second language (possibly the 'young adults' PIS could be adapted for use with this group).

12. Provide an audio/video of the content of the PIS and consent form for participants who find access to the written word difficult.
13. Promote the Genomics England website as a place to access information (presented in a variety of formats) about taking part in the project.
14. All materials to adopt consistent use of terms relating to anonymisation, when it is linked-data, when unlinked and clarify at which point this occurs e.g. at what point companies might have access.
15. More information to be available to participants regarding the concept of additional findings and especially ones of reproductive significance, including the timescale for results and future contact.
16. To clarify the audience for the tri-fold information booklet and if it is for use with patients, review the booklet and add definitions of terminology used.

**Changes to consider - Process of Consent Interaction (17-22)**

17. Explore ways of ensuring that recruiters have appropriate time to discuss the issues raised in the PIS with participants, where this is needed.
18. Support recruiters through on-going consent training including how to manage questions from participants regarding support with communication in their families and social/psychological support.
19. Support recruiters with the information technology systems for registering participants and logging samples.
20. Provision of more publicity materials to help recruitment into the project.
21. A staged consent process for cancer participants if anxiety levels are high at the time of diagnosis. Possibly consider adapting the PIS and consent forms for this patient group.
22. During the further integration of whole genome sequencing into routine NHS clinical care, the following issues need careful consideration and planning.
  - 22.1 Maintaining adequate time for consent discussion.
  - 22.2 Consider carefully the changes required for the transition of Whole Genome Sequencing consent from the research environment into routine clinical care.
  - 22.3 Supportive and appropriate local mechanisms for receipt of main findings and additional findings (results).
  - 22.4 Support and updating of staff providing patient support.
  - 22.5 Consideration of consent timing within the routine care pathway (especially cancer patients and their families).

Figures 3 Infographic Summary of Evaluation

## NHS GMC: National Service Evaluation of Consent Process and Participant Materials used in the 100,000 Genomes Project

### Snap-shot Survey

Eight week period  
early 2016



1196 100,000 GENOMES PROJECT PARTICIPANTS CONSENTED

14.6% Participants gave feedback

161 ACTIVE RECRUITERS  
36% gave feedback

5 PPI FOCUS GROUPS, 34 MEMBERS  
100% gave feedback

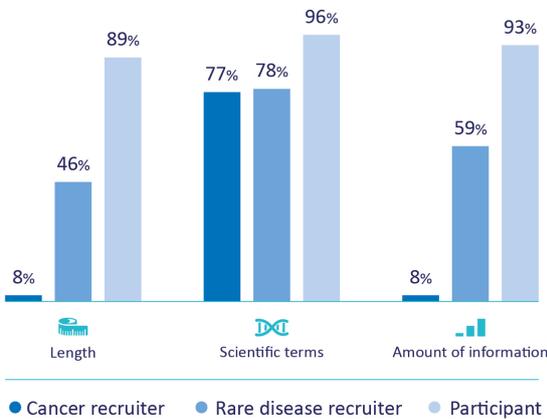
### FEEDBACK DIFFERING VIEWS



98% of participants described their consent experience as GOOD or EXCELLENT

Focus Group Members and Recruiters felt the Participant Information Sheet and consent forms needed revision

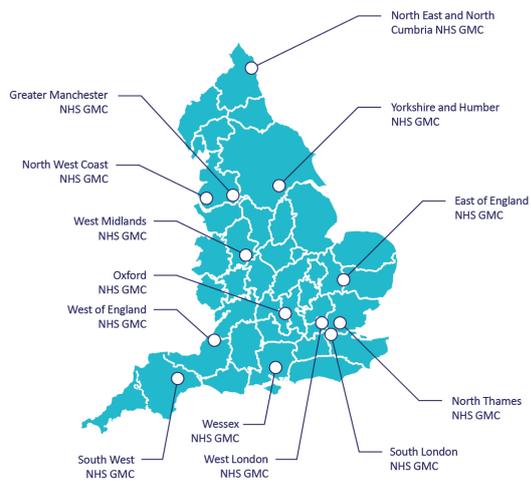
### COMPARISON OF PERCENTAGE OF PARTICIPANTS & RECRUITERS ON WHAT IS "ABOUT RIGHT" WITH PARTICIPANT INFORMATION SHEET



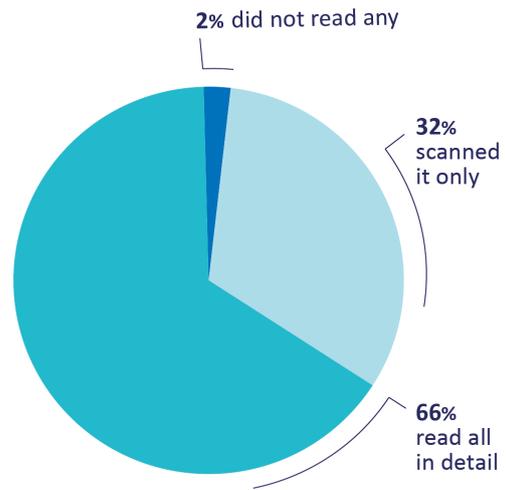
### NHS Genomic Medicine Centres Paving the way to personalised medicine



The Evaluation involved all 13 GMCs



### HOW MUCH PARTICIPANT INFORMATION SHEET DID YOU READ?



50%

of recruiters said their participants OFTEN or VERY OFTEN presented with specific needs



Needing extra explanation



Low levels of literacy

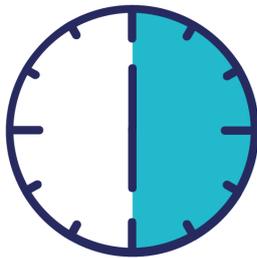


Visual impairment



High level of anxiety

THE PROCESS



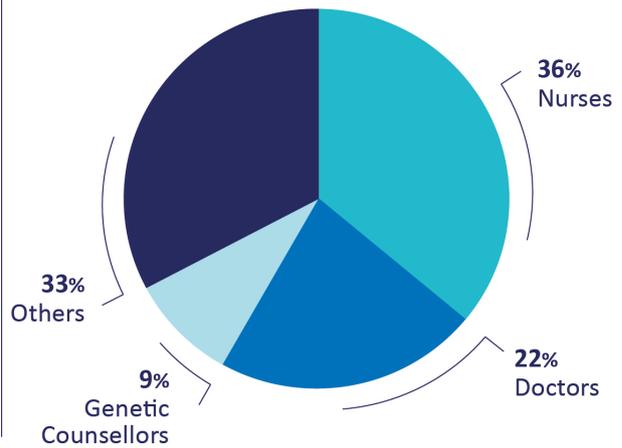
RARE DISEASE CONSENTS

19% <30MIN  
<25% IN ROUTINE NHS CARE

CANCER CONSENTS

45% <30MIN  
>75% IN ROUTINE NHS CARE

WHO IS TAKING CONSENT?



THINGS TO KEEP, THINGS TO CONSIDER CHANGING.

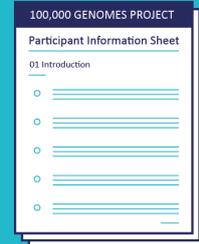
23 RECOMMENDATIONS

- Improve the presentation and readability of the Participant Information Sheet and consent forms.
- Support recruiters with on-going training and explore ways of ensuring appropriate time for consent discussion.
- Improve access to all consenting material e.g. through sign-posting to easy-read /visual materials on the website.
- Ensure clarity and consistency of use of project and scientific terminology.
- Consider changing the consent process for cancer participants.

REGULATIONS | POLICY



REVISION OF CONSENT MATERIALS AND PROCESS



RESEARCH EVIDENCE

## 1 Background

### 1.1 Context of Consent in the 100,000 Genomes Project

In April 2015, the first set of consent materials were issued to Genomic Medicine Centres. The materials and consent process had been developed by Genomics England after two years of piloting, policy and clinical discussion, including engagement and involvement activities with patients, clinicians and other groups. Some of the key engagement and advice reports for the period prior to 2015 are detailed below and are available on the Genomics England website.

- Advisory Group on Ethics letter to Dame Sally Davies (Chief Medical Officer) 22<sup>nd</sup> March 2013 - Professor Michael Parker (Chair of the advisory group on ethics).
- *Managing incidental and pertinent findings from Whole Genome Sequencing (WGS) in the 100,000 Genome Project*. A discussion paper from the Public Health Genomics Foundation. April 2013.
- *Potential Participant Literature: Research Report*, November 2014. RandallFox.
- *What do patients with rare genetic conditions think about whole genome sequencing in the NHS? Research Findings for the 100,000 Genomes Project*, November 2014. Genetic Alliance UK.
- *Ethical issues relating to involvement of cancer patients in the 100,000 Genomes Project. Qualitative Research Findings*, October 2014. GfK (Market Research Company).
- *Research on Genomic Data Sharing. Qualitative research report*, September 2014. Solutions Research (Market Research Company).

The consent materials and process also had to satisfy legislation such as the *Data Protection Act (1998)* and the *Human Tissue Act (2004)*, as well as the *Mental Health Act (2007) (Department of Health 2007)*, in order to provide for consent to be sought for certain uses of tissue or data.

The 100,000 Genomes Project is unusual in that it is not purely a research study, but is embedded in the patient's own clinical care team, issuing results back to participants and their families. Therefore the consent process and documentation had to be designed to work to inform participants about both the clinical utility and the research aspects of participation.

*"Drawing on relevant guidance and exemplars of good practice, the participant materials (i.e. letters of invitation, Patient Information Sheets, and assent and consent forms) have been developed in accordance with the key principles in the Key Ethical Principles document and Ethical Governance Framework, and with the advice of the Ethics Advisory Committee, with comments from the Science Committee, and other stakeholders and the appropriate guidance."*

100,000 Genomes Protocol, February 2015.

### 1.2 Context of the National Service Evaluation Project

Some NHS Genomic Medicine Centre (GMC) Patient & Public Involvement (PPI) groups commented on the consent documentation which was issued to GMCs at the start of the project (April 2015). They suggested that it would benefit from an evidenced review, which this service evaluation aimed to provide.

Genomics England set out a phased framework for the evaluation and adaptation of the consent documentation and process. Phase 1 consisted of seeking feedback from PPI leads and clinicians within the NHS GMCs as recruitment began. This resulted in the development of additional items of participant information materials, for optional use, which aim to respond to (potential) participants' different needs. The following amendments and additions were made to the consent materials between April 2015 and March 2016:

- A short 4 page introduction leaflet (executive summary)
- A tri-fold awareness-raising pamphlet
- Editing of the Research Ethics Committee (REC) approved consent materials to correct mistakes and inconsistencies
- A Prezi presentation for participants, as an alternative to print media
- Materials for partially sighted and blind participants
- Flip chart for use during consent discussions

### **1.3 Shared purpose and scope of project**

Genomics England engaged in discussions regarding how to gather evidence which could be presented to the Research Ethics Committee (REC) alongside any suggested changes.

In August 2015 Genomics England created an Evaluation Project Steering Group, which reviewed an outline proposal led by North West Coast GMC in collaboration with two GMC PPI leads. This described an evaluation which would provide the opportunity for receiving feedback from three stakeholder groups (participants, recruiters and GMC PPI members). The project team and Genomics England staff would work together, using the NHS Change Model of the NHS Improving Quality programme as a framework to identify a shared understanding as to the reason for the evaluation, as well as the process by which results would feed back into change to the consent materials or process (NHS Improving Quality 2015).

In March 2016 the evaluation project schedule was agreed, (<https://www.genomicsengland.co.uk/consent-evaluation>) and was approved by the REC on 18<sup>th</sup> May 2016 (14/EE/1112 Substantial Amendment 8). A commitment to evaluate the consent materials was outlined in section 12.2.1 of the 100,000 Genomes Project approved Protocol v 2.0, dated February 2015 (Available from <https://www.genomicsengland.co.uk/library-and-resources/>). As such this schedule is in accordance with the Protocol v 2.0 as approved by the NRES REC, and requires no change to be made to this.

A commitment was also made to publish a public report outlining the findings of this evaluation, alongside other evidence, to make recommendations concerning any amendments required.

The Genomics England Steering Group agreed part-funding, alongside funding offered by the Innovation Agency (NWC) and a collaboration partnership between the University of Central Lancashire, University of Birmingham and Imperial College London.

The NHS Genomic Medicine Centres' National Service Evaluation of the Consent Process and Participant Materials used in the 100,000 Genomes Project had three aims:

1. To evaluate participant materials (consent documentation)
2. To evaluate the consent discussion process
3. To provide evidence to inform the development of consent materials and processes for the NHS in this area

The service evaluation covers three key stakeholder groups:

- NHS GMC PPI group members
- Healthcare professionals (recruiters), and
- 100,000 Genomes Project participants

The aim being that views could be collated and any problems resolved for the benefit of future participants, although re-design of the consent materials is beyond the remit of this service evaluation.

The results of this research project have been submitted to the Genomics England Ethics Advisory Committee. This independent committee advises the Genomics England Board on the ethical aspects of everything Genomics England does. It scrutinises what information patients should receive about their results, as well as the policies on consent. This work forms part of Genomics England's wider evaluation of the 100,000 Genomes Project consent documentation and consent discussion process.

## 2 Methods

### 2.1 Evaluation Methodology

This project is a developmental evaluation, defined as:

"Where innovation assumptions are revised over time, with the results that the goal of the intervention may be changed. This type of formative evaluation also facilitates close to real-time feedback to the intervention team. It assists with trying out new ideas, documenting activities and their short-term consequences, identifying processes and outcomes as they emerge and helping people make sense of them" (Dozois, Langlois et al. 2010).

The evaluation was a mixed methods service evaluation and used a triangulation approach for gathering evidence (Creswell 2011).

The design is parallel multi-strand mixed methods. In this design the qualitative and quantitative phases (activities) were designed to answer related aspects of the same research question (Teddlie 2009). "In this design the strands of analysis are independent, each providing an understanding of the phenomenon under investigation. These inferences made on the basis of the results from each strand are integrated or synthesised to form meta-inferences" p 266 (Teddlie 2009). One advantage of mixed methods research is that of providing an assortment of divergent views (Greene 2007). The parallel tracks analysis was undertaken according to the markers of quality and excellence for each method (qualitative or quantitative) (Figure 4).

Specific areas of interest covered both the rare disease and cancer 100,000 Genomes Project pathways, and included:

- Consent discussion process - the context within which the discussion occurs.

- Are participants given appropriate time to read information about the project and discuss with others before making their decision to participate?
- How long does the consent interview last?
- What facilities are available to the participant and health professional?
- Participant information sheets – content, visual impact, layout, logical order/flow of topics.
- Participant consent forms – content, visual impact, layout, logical order/flow of topics.
- Health Professional Support.
  - Are the recruiting health professionals supported in providing the participant with an explanation of the project and answering participant questions?
  - How easy or difficult do they find consenting participants for this project?

Working with three different stakeholder groups has permitted evaluation of the consent experience in multiple geographic sites and from contrasting perspectives. Materials for data collection were piloted and field tested for face validity and were amended appropriately. It was anticipated that the materials would be used by all NHS GMC sites to feed data back to the Service Evaluation Project Team for analysis and synthesis. Each site had the option to include either or both rare disease and cancer participants.

**The intervention being evaluated:** The 100,000 Genomes Project consent materials and consent discussion process in use February to July 2016.

The project started with 10 pre-determined areas of focus, divided into two main categories. The areas of focus were extrapolated by the Project Team from their discussions with the Steering Group, the NHS GMC PPI group network, the EAC, NHSE, and other wider stakeholders.

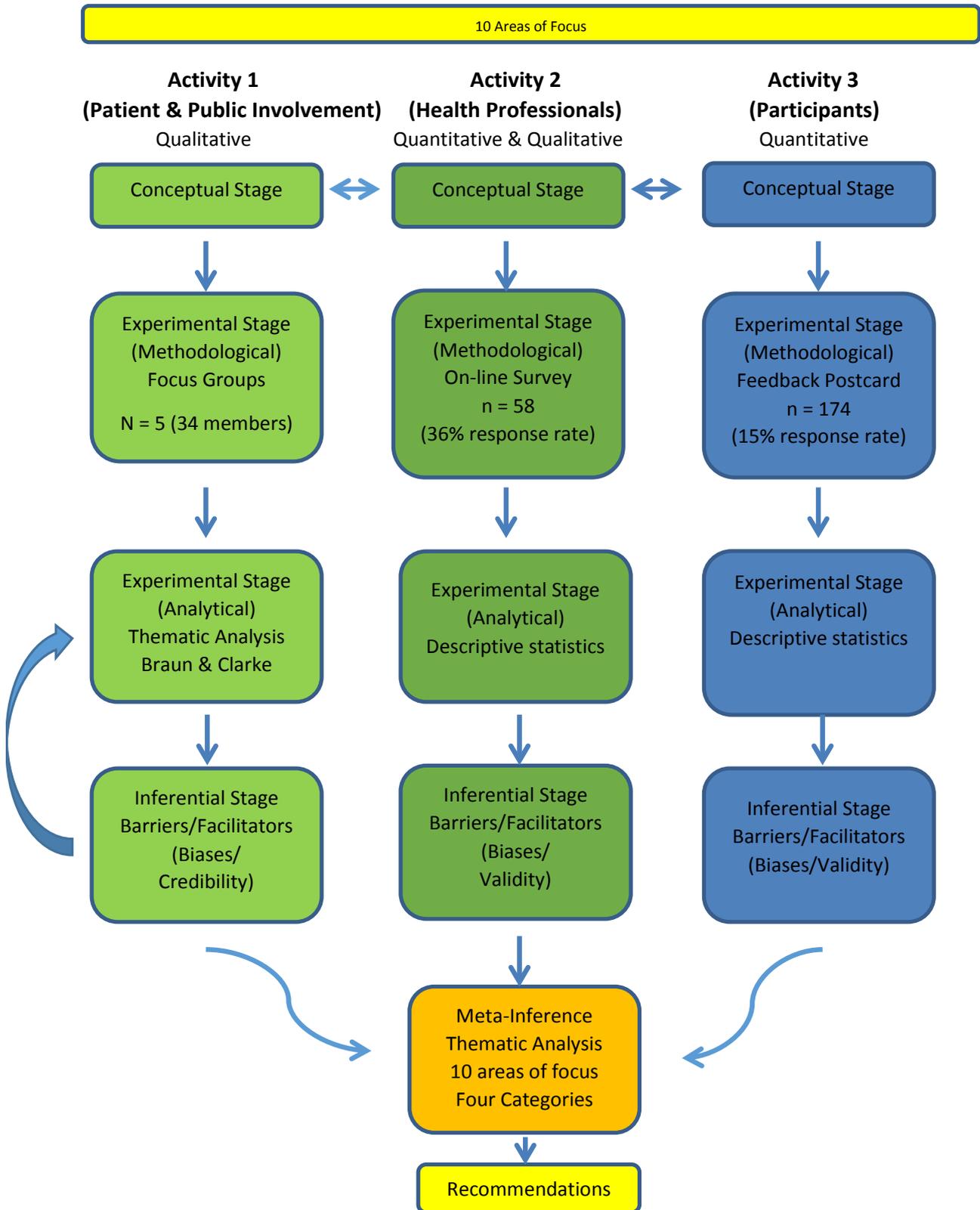
#### **Project Documentation**

1. Logical Flow
2. Complexity of project structure (complex project – consent material is therefore complex)
3. Complexity and consistency of use of scientific/genomic terminology
4. Issues related to main findings, additional (opt in/opt out) and incidental findings
5. Data security (privacy) and data sharing (private companies) – including impact on insurance

#### **Process of Consent Interaction**

6. Nature and timing of approach to participant – (personal/letter)
7. Seeking broad vs. specific consent
8. Time taken for participant to consent (at initial contact, after several contacts)
9. Consent Interview – location/length/ease
10. Flexibility vs. pre-determined pathway

Figure 4 Project Design - Parallel multi-stand mixed methods design



## 2.2 Focus Group Methods

The NHS GMC PPI groups' focus group activity aimed to elicit feedback on consent material, process, and ease of use of the current participant documentation in place during the snap-shot survey time period.

The focus group enquiry concentrated on the following four topic areas:

- Topic 1. Logical Flow of consent documentation
- Topic 2. Complexity of project structure
- Topic 3. Seeking broad consent for additional findings
- Topic 4. Transformation and sustainability

NHS England contacted the NHS GMC Clinical Directors and they, in collaboration with the NHS GMC PPI Leads Network, nominated a local contact, a nominated PPI lead, for their NHS GMC. The nominated PPI leads collaborated with NHS England on how best to facilitate convening the focus groups with GMC PPI group members. Criteria for focus group membership are detailed in the research schedule document. The research team asked local PPI leads to send invitations to individuals who had prior knowledge of the 100,000 Genomes Project, who were not necessarily participants, but ideally had worked with the GMC providing PPI input. An evaluation project participant information sheet and a consent form together with the 100,000 Genomes Project participant material were sent to potential participants prior to the focus groups. Prior to commencing the focus group, the consent forms were collected together with a short socio-demographic questionnaire. Focus group participants were provided with travel expenses for attending the meeting, in line with INVOLVE guidelines; refreshments and lunch was also provided. A 'thank-you' voucher (£25) was also sent after the event to each member.

All focus groups were digitally recorded, subject to the participants completing the written consent form. Recordings were professionally transcribed. Data derived was grouped thematically within the four topic areas above and, where relevant, the other pre-existing areas which have been identified from previous patient and public research concerning the consent materials.

Data was analysed by Markella Boudioni at Imperial College London using the qualitative methodology of thematic analysis (Braun and Clarke 2006). This is a qualitative method aiming to answer the evaluation questions and is not a theoretically based qualitative analysis. The documents that were distributed to participants can be found in the project Schedule document available from <https://www.genomicsengland.co.uk/consent-evaluation/>.

## 2.3 Health Professional Survey Methods

The aim of the survey was to elicit the opinions of NHS GMC health professionals who are actively recruiting participants, and focused on the consent materials and consent discussion process in place during the February to March 2016 time period.

NHS England contacted the Clinical Director of each NHS GMC and asked them to nominate a member of their GMC who would act as a local contact for the health professional recruiter survey. The NHS GMCs sent an invitation email to their recruiters, including an evaluation project participant information sheet for health professionals and a link to the anonymous survey, which was protected

by a generic password. The GMCs supplied the number of recruiting health professionals to whom they sent the survey link.

The survey questions were piloted and amended accordingly. The survey contained closed and open ended questions and took about 10-15 minutes to complete. The quantitative questions were analysed by descriptive statistics using the SPSS package (IBM Corp 2013). Where possible, responses from open text questions were grouped by themes or are provided verbatim. Data-analysis of the health professional survey occurred independently of the analysis of the focus groups, but was concurrent with analysis of the participant survey.

The health professional survey, introductory email, participant information sheet and survey questions can be found in the project Schedule document

## **2.4 Participant Feedback Methods**

The Clinical Director of each GMC was asked to name a local contact within the NHS GMC who would be able to facilitate the distribution of participant survey postcards to recruiting health professionals.

Each NHS GMC was asked to estimate how many participants they expected to consent during the 2month evaluation period. The NHS England team supplied each GMC with a pack containing two sets (rare disease participants and cancer participants) of numbered postage paid survey postcards (Figure 5), which had a unique postcard number linked to their NHS GMC. This number does not link to the patient's clinical or 100,000 Genomes Project identifiers. The participant survey was anonymous.

The recruiting health professionals were provided with briefing notes and asked to explain the survey to the 100,000 Genomes Project participants for whom they countersigned a consent form. The participant was asked if they wish to take part in the survey. If they did, they were handed a survey postcard before leaving their consent interview.

The aim of this postcard distribution strategy was to calculate a response rate based on the number of signed consents uploaded to the OpenClinica system over the period (monitored by Genomics England) vs. the number of postcards returned by participants during the snap shot survey date window.

The postcard stated the aim of the evaluation and confirmed that all feedback is anonymous. The reference number on the postcard refers to the centre recruiting the individual; the individual survey number does not link back to any personal identification. Participant information about the Service Evaluation was printed on the postcard and was also explained verbally by the health professional presenting the postcard. Consent was inferred by the participant returning the postcard.

Participants were also given the option to complete the survey on-line by entering their unique postcard number. The question set was identical on the on-line option, apart from one additional question which allows the participant to provide more open comments.

To optimise accessibility and inclusion the website option facilitated options such as large font and could also be used with text readers. The postcards were returned to the Service Evaluation Project Team for analysis.

The survey contained 15 closed and one open question and took about 3-5 minutes to complete. The quantitative questions were analysed by descriptive statistics using the SPSS package (IBM Corp 2013). Where possible, responses from open text questions were grouped by themes or are provided verbatim. Data-analysis of the participant survey occurred independently of the focus groups, but was concurrent with the health professional survey.

The documents that were distributed to participants can be found in the project Schedule document.

Figure 5 Participant Postcard – Suspected Cancer (C card) and Rare Disease (R card) Participants



**Genomics**  
england



**Genomics**  
england

**Freepost GENOMICS ENGLAND**

**National Evaluation - Thank you for taking part**

**PARTICIPANT FEEDBACK CARD**

**We would like to know your opinion of the 100,000 Genomes Project consent process**

By completion of this card I give consent to give my views to this survey anonymously. You can also leave feedback at [www.genomicsengland.co.uk/consent-evaluation](http://www.genomicsengland.co.uk/consent-evaluation) and entering the number on this card.

**Accessible options for feedback are available online.**

**AFTER COMPLETION PLEASE FREEPPOST in any post box.**  
These feedback cards are numbered by NHS Hospital. All responses are fully anonymous and not linked to any personal identification details.

**Ref No.**

**Please tell us what you think**

**Please tick which of the following project materials you read or viewed prior to consenting to participation in the 100,000 Genomes Project.**

Participant Information Sheet & Consent Form  Flip Chart   
 Introductory Booklet  Videos or Animations   
 Consultee Information Sheet & Consent Form

How do you rate the overall satisfaction with your consent experience?  Excellent   
 How do you rate the written information given to you about the project?  Good   
 How do you rate the verbal information given to you about the project?  Satisfactory   
 How do you rate the care and attention you received during the process?  Poor

**How much of the participant or consultee information sheet did you read?**

Read all in detail  Scanned all but did not read in detail  Did not read any

**What did you think about the participant or consultee information sheet you were given to make an informed choice?**

**Length**  
 Too Long  Too Short   
 About Right Length

**Scientific Terms & Words**  
 Too Simple  Too Complicated   
 About Right Level

**Amount of information**  
 Too Little Information  Too Much Information   
 About Right Amount

**In the information sheet would you have liked more or less detail to be included about:**

Use and storage of my samples and health data  Less  More   
 Results related to my main condition  Right   
 Additional results unrelated to my main condition  Less  More

**Did you feel you were given enough time to understand all the information prior to signing the consent form?** Yes  No

**Please indicate what category of participant you are:**  
 An adult patient with cancer (or suspected cancer)   
 A personal consultee

Gender: Male  Female  Other   
 Age Group: 16-24  25-44  45-64  65-74  75+

**Would you have been satisfied with a shortened information sheet as the complete text is available online?**  
 Yes  Maybe  No

**Were there any bits (documents or process) of the consent experience that were difficult or confusing for you or your family?**  
 .....



**R**  
Ref No.



## Freepost GENOMICS ENGLAND

### PARTICIPANT FEEDBACK CARD

We would like to know your opinion of the 100,000 Genomes Project consent process

By completion of this card I give consent to give my views to this survey anonymously. You can also leave feedback at [www.genomicsengland.co.uk/consent-evaluation](http://www.genomicsengland.co.uk/consent-evaluation) and entering the number on this card.

**Accessible options for feedback are available online.**

**AFTER COMPLETION PLEASE FREEPOST** in any post box.

These feedback cards are numbered by NHS Hospital. All responses are fully anonymous and not linked to any personal identification details.

### National Evaluation - Thank you for taking part

Please tell us what you think

Please tick which of the following project materials you read or viewed prior to consenting to participation in the 100,000 Genomes Project.

- Participant Information Sheet & Consent Form  Flip Chart
  - Introductory Booklet  Videos or Animations
  - Consultee Information Sheet & Consent Form
- How do you rate the overall satisfaction with your consent experience?  Excellent
- How do you rate the written information given to you about the project?  Good
- How do you rate the verbal information given to you about the project?  Satisfactory
- How do you rate the care and attention you received during the process?  Poor

How much of the *participant or consultee information sheet* did you read?

- Read all in detail
- Scanned all but did not read in detail
- Did not read any

What did you think about the participant or consultee information sheet you were given to make an informed choice?

- Length**
- Too Long
- About Right Length
- Too Short
- Scientific Terms & Words**
- Too Simple
- About Right Level
- Too Complicated
- Amount of Information**
- Too Little Information
- About Right Amount
- Too Much Information

In the information sheet would you have liked more or less detail to be included about:

- Use and storage of my samples and health data  Less  Right  More
- Results related to my main condition  Less  Right  More
- Additional results unrelated to my main condition  Less  Right  More

Did you feel you were given enough time to understand all the information prior to signing the consent form? Yes  No

Please indicate what category of participant you are:

- An adult patient with a rare disease
- An adult family member of a patient with a rare genetic disease
- The parent(s) of a participating child
- The parent(s) of a deceased child
- The nominated representative, friend or personal consultee
- Gender: Male  Female  Other
- Age Group: 16-24  25-44  45-64  65-74  75+

Would you have been satisfied with a shortened information sheet as the complete text is available online? Yes  Maybe  No

Were there any bits (documents or process) of the consent experience that were difficult or confusing for you or your family?

.....

## 3 Results

This section presents the results from each stakeholder group, these results are then combined and presented in Chapter 4, Synthesis.

### 3.1 PPI Focus Groups

Basic demographics are presented followed by results of the four topic areas and additional barriers and facilitators.

#### **Description & socio-demographics**

Five focus groups of 5-8 members took place during May/July 2016 at various locations linked with ten out of the thirteen GMCs, and covered rural and urban localities.

The characteristics of the 34 focus group members varied. There were groups with 100,000 Genomes Project participants, members of patient groups - specific or non-specific to the project - and mixed groups. One group was conducted with patients and relatives/carers of people with rare diseases; all other groups included both patients with cancer, rare conditions, and relatives/carers of people with rare diseases. The majority were males and females between 45-64 and 65-74 years old, with only two groups including younger adults aged 25-44 years old. Members resided in a variety of locations; they were mainly White British with the exception of London members.

#### **Experience of the 100,000 Genomes Project**

*The experience of Focus Group members varied:* One group consisted of 100,000 Genomes Project participants/patients with rare conditions and their relatives. Four groups were mixed condition groups involving Project participants and members of other groups; one group did not involve any Project participants.

*Participants of the 100,000 Genomes Project:* A few of the 100,000 Genomes Project participants with rare conditions were also involved with other projects, such as Biobanking and Tissue Banking; they suggested the Project had a clearer remit and benefits than other projects (FG A, FG E).

*100,000 Genomes National Participant Panel:* A few focus group members were members of the Panel. They were critical about its first meeting; the contribution/expectations from them were unclear (FG A, FG E). However, they were optimistic as they could see a 'physical research goal' (FG A).

#### **Four topic areas for Focus Group enquiry**

Topic 1. Logical flow of consent documentation

Topic 2. Complexity of project structure

Topic 3. Seeking broad consent for additional findings

Topic 4. General transformational and sustainability issues

### Topic area 1 - Logical flow of consent documentation

#### **General**

There were differing opinions about the consent material, but few members found the material adequate. Consensus that it was adequate and 'it covered what it should cover', was reached in only

one group (FG A); members felt that the format and the layout of the information sheets and consent forms with the different colours, bold letters, boxes and key points were helpful in highlighting specific issues. Members of other groups liked the '*clean*' presentation and colours (FG B, FG E), the subtitles in a question format, the key points in boxes of the information sheets, and boxes in the consent forms (FG B). Interestingly, those with positive opinions about the consent material were mostly 100,000 Genomes Project participants.

Within the other groups, some members thought the material was appropriate, good and fit for purpose (FG D); that people may want different types and level of information, thus difficult to provide for everyone (FG C). One member said '*it strikes a good balance managing expectations*' (FG E).

Other members thought the information in general was too wordy (FG A, FG B, FG D), dense and dry, with a lot of jargon and inaccessible language (FG A, FG C): '*they are typical medical research leaflets and feel cold*' (FG D). Members also found the material not '*user friendly*', and not meeting the 9 or 13 year old literature standards (FG C, FG E). Concerns about the forms' design, layout and style were also raised; some thought the angle of the documents was rather negative and not selling the benefits of the project (FG D, FG E). Another suggested a shift from '*creating a legacy*' to '*why am I doing this*' and personal and family benefits (FG C). In one group there was a consensus that the overall emphasis should be on early detection, prevention, treatment and management (FG D).

### **Participant Information Sheets (PIS)**

There were differing opinions about the PIS, discussed in all focus groups, but only a few members were satisfied with the flow of information. Members in one group said the forms were clearly laid out and no more difficult than many other official forms (FG E); two members of another thought the information and its presentation '*covers its purpose*' (FG D).

The majority of members had a number of comments: the PIS were too wordy, lengthy (FG B, FG C) or information dense (FG E). It was recognised however that there is no easy answer to this, as consent needs to be fully informed (FG E). Potential problems and difficulties in understanding for most English speakers (FG E), but also for non-native English speakers (FG C) were discussed.

Members highlighted a gap about referring to emotional support, regarding both available support and the process (FG D). Members also suggested further explanations and clarifications of scientific/genomic language were needed, i.e. sequencing (FG A, FG B, FG D, FG E). Many other suggestions about language and more accessible/lay explanations were made (FG B, FG D, FG E), and are detailed in the 'National Evaluation Project: Background Document' available from the Project team or Genomics England on request.

### **Consent forms**

Some members in one group liked that the consent forms were broken down clearly into different sections to sign (FG D). But another group thought the information included was not sufficient, not easy to read or understand (FG E). Although there were fewer comments in general about the consent forms, a few language and editing suggestions were made (FG D, FG E).

### **Opt-in and Opt-out forms**

Some language suggestions were also made (FG D). Members said that '*additional findings*' should be explained so people knew what they were consenting to.

## Suggestions

Suggestions included simplified language, i.e. clear plain English (FG A, FG B, FG D), and accessible, easy to understand information (FG A, FG B), with the use of more visual material - pictures, charts and diagrams - (FG A, FG B), e.g. a diagram with the pros/cons of each topic (FG B). Some examples would be useful to provide context, such as the likelihood of additional findings (FG E). As visual material would not be suitable for those with visual impairments, other formats should be developed (FG A).

All 'jargon' should be removed, and clarifications may be needed to make the material more user friendly, i.e. the description of 'genomic medicine' with an example (FG C). Clarification and explanation of scientific/genomic language, i.e. 'genome' and 'genomic medicine', and consistency across the different documents was suggested (FG A, FG B, FGD), together with more explanation of other terms (i.e. clinical trials, carrier status) (FG B, FG E). These could be explained further in the text or with a box at the end of the PIS (FG B). Alternatively, an extra leaflet could define these terms, including description and explanation of other bodies/structures pertinent to the research, such as 'teams' (FG C). The addition of a glossary was strongly suggested by most groups (FG A, FG B, FG C, FG E).

The language could be changed positively to sell what the project would do in future and its benefits, perhaps adding success stories and a paragraph about emotional support (FG A, FG D).

Figure 6 Logical flow of documentation

*'I think, for what they are, the form [PIS] is quite clearly laid out. At the end of the day to take part in the project you have to tick all of these boxes and you can't argue or seek exemption on this element or that element....'*

**GMC PPI Group member, parent of child with rare disease, non-participant**

*'...I didn't think the language was very accessible...I think given that it was an introduction out of nowhere... It wasn't in clear English...I think it could have been written in more plain English. It was a bit dry as well so it wasn't appealing...I think this was also selling us something, if you like, but it didn't do a great job of it because it already wanted to buy.'*

**100,000 Genomes Project participant, patient with rare disease**

*'I really think if you've got a leaflet, of some sort, that gives it in an easy read form, and you'd probably read it yourself, 'now I understand and now I'll go onto these'. To me it'd be a welcoming thing, 'Don't be put off by all this paperwork. We are obliged to give you this information', and spell it all out in a lighter format and then have the forms.'*

**GMC PPI Group member, patient with cancer, non-participant**

## Topic area 2 - Complexity of project structure

### General

In general, it was recognised as a complex project (FG A, FG C, FG E) and genome exploration as a difficult concept to understand (FG E). The complexity of all material, clarity, consistency of language, and cross-referencing presented earlier also add to the complexity. Other elements were highlighted, i.e. changing your mind later (FG B), the availability of genetic testing anyway (FG E).

Only a few members felt the project is quite straightforward (FG C, FG E), 'the project itself is fairly simple, it's the consequences that have the complexity' (FG C).

### Rare conditions and cancer

The differences between rare conditions and cancer were highlighted, e.g. in terms of time scale of diagnosis (FG E), the associated differences in the consent process and timing in the patient pathway

(FG B, FG D, FG E). Patients with cancer and rare conditions and their families may have different views of the Project (FG B, FG E); they need different kind of counselling and follow up (FG E). Those with rare conditions have potentially more to gain from this work (FG E); the need to find a cure/help their families was recognised (FG A, FG B, FG E).

### **Consent process – 100,000 Genomes Project participants**

For participants, the consent process was variable, depending on the Trust, locality and condition (FG A). For some people with rare conditions, the process was smooth and fast; the majority received information by post, a few were contacted by phone first, followed by an appointment (FG A). Other people, either with cancer or rare condition, received a clear simple invitation email (FG B). Others were told their eligibility by their clinician and then received the information by post (FG A). One participant mentioned an open day for all patients who could potentially be recruited (FG C). Examples of good practice included professionals explaining the information verbally and spending a lot of time with participants (FG A, FG E) - including people with dyslexia and visual impairment conditions (FG A).

### **Consent process – Non-participants of the 100,000 Genomes Project**

For non-participants, it was unclear what the consent process entailed (FG C, FG E), if people could be self-referred or were referred by their clinicians (FG E). They thought the project would generate many questions, i.e. about the impact on treatment, and involvement of family members (FG E). It was recognised as a complex project that would require explanations by a health professional (FG E).

One focus group member raised concerns about the process and the point of their pathway at which patients were asked to participate; he cited examples when people were asked to participate *'within 5 minutes of a breast cancer diagnosis'*, urging caution in sensitive areas (FG C).

### **Complexities regarding carrier status, additional findings and mental capacity**

It was acknowledged that anything with the potential to involve others requires *'extra thought'*, that people are individuals and want to know different things (FG B). Someone questioned the extent of *'knowing'* in terms of likelihood of transmission of a disease (FG B). Relevant to this were issues about mental capacity; a member, who was also a 100,000 Genomes project participant, brought the example of his elderly mother who was unable to consent, but he had lasting power of attorney. It was unclear how these cases are handled (FG A).

### **Suggestions**

The importance of the interaction, professionalism and competency of professionals taking consent was stressed (FG C, FG E). Recruitment should be reassuring; the onus should be on recruiters having enough time to provide appropriate information (FG C). Written information was considered essential, so people can take it away; but the project would need explaining in person by a health professional as most people would prefer to listen rather than going through the written information (FG C, FG E).

Members were not clear that the forms are available online through the 100,000 Genomes website. Some members would like to be able to read the information and sign the consent form online and not through their clinical teams (FG A).

The availability of support, counselling or *'tips or advice'* for how to broach this topic or break such news to affected family members was discussed (FG B, FG D). There should be strong recommendations to discuss this with family members at the point of PIS. Support should be made available at a local level; the support process and level should be made clear to potential

participants (FG B, FG D). It was suggested that this is likely to increase participation and retention (FG B).

In addition, more emphasis should be placed on explaining data security issues and clarifying the benefits of participating to potential participants (FG C, FG D).

Figure 7 Complexity of project structure

*'...it's quite a difficult concept for people to understand because it's not a classic health service thing where if you've got X you go Y and you get some treatment. It's a different mind-set...it is a different philosophical mind set. I think it's very different.'* **GMC PPI Group member, patient with cancer, non-participant**

*'... some of the participants in this project, it is the only way - or only avenue they have to find the name or a potential cure for their disease either for them or their family...Because people feel a duty to their sons, their daughters, their granddaughters.'* **GMC PPI Group member, unspecified condition, non-participant**

*'...So if you're given a result that you're a carrier, if you are given a result that you may be prone to a cancer in the future, there's no mention here of any support you might be given to understand or to support you through the emotional difficulties that you may get from those results. I think that is very important.'*

**Patient Group member linked with GMC, patient with rare disease, non-participant**

### Topic area 3 - Seeking broad consent for additional findings

#### General

Opinions were variable about additional findings; people are individuals and want different things (FG A). More information was suggested, i.e. what additional findings are, examples and glossary (FG E).

#### Online availability of opt-in and opt-out additional findings forms

It was unclear if the forms were also available through clinical teams (FG B, FG D). Thus, there were concerns about them being available online only and potential drawbacks, i.e. equipment, technology, time, and reduced access for many (FG B, FG D). Someone questioned the clinical teams' ability to provide forms and suggested a downloadable form to be retained (FG D). Other suggestions were individual GMCs being made responsible for the collection and retention of this information, i.e. through GMC nurses (FG B, FG D); another was a freepost service to Genomics England (FG B).

The majority would like easy access to both online and paper versions (FG C, FG D); paper versions could be given with the participation pack (FG C). Most members of one group agreed that some people might make a rash decision; having the option of changing one's mind in a quick and easy way such as downloading/sending off a form *'wouldn't be conducive to a rational and well considered choice'* (FG C). One member suggested an online *'request form'* and not a download; two others described this as *'too convenient'*; they would welcome a consultation around opting-in or out (FG C).

#### Information about additional conditions available online

In three groups, members felt - as additional findings are not clear - unsure about how to answer the question posed in the group about information being available online (FG A, FG C, FG E). They were confused about whether the list of additional findings was fixed, or updated (FG E). They did not feel knowledgeable enough to make clear suggestions (FG A).

Responses of those who would like to have information was variable; most found the online list of additional conditions sufficient (FG C). Others would like to have *'all'* information available online, with links to further information (FG A), and information about the *'added value'* of additional findings (FG D). Others would like to know what additional findings have been found, the percentages linked with other conditions (FG C), potential trends and geographical variations (FG D).

Figure 8 Current process: Opting-in or out of receiving additional findings

*'I'm one of those people who just wants it all, I would love it...So I say, 'put it all out there', but don't throw it in people's faces. Have the links there for the people who want to trawl through and drill down... it would be important to also make sure that there were adequate links about where the information is going to be, basically who might buy it, who might be using it, and what they might be doing too. If they're going to have access to the information then they jolly well need to put something about commercial organisations who are working with the data...'*

**100,000 Genomes Project participant, patient with rare disease**

*'There is still this element of patient choice. You cannot take away patient choice, it is fundamental thing of our statutory rights, you have to have choice. You can't just take it away from people.'*

**Patient group member linked with GMC, patient with rare disease, non-participant**

The majority of members would like the choice of opting-in or out of their original consent decision (FG A, FG C, FG D, FG E); they also liked the option of doing it online. In two groups, there was strong agreement on this (FG D, FG E); *'people need to understand what they sign up to when they opt in'*, as it has important implications (FG E) and *'patient choice'* is an element of the NHS (FG D).

In one group, and if they had one option only, members reached a consensus after a long discussion for opting-in for additional findings (FG B). For some members of other groups opting-in was also the preferred option (FG A, FG C, FG E); a member felt that one should always be presented with the option of opt-in, so people are conscious about what they are consenting to (FG C). Only one member felt that opting-out was better, as it would be best for recruitment (FG C).

## Topic area 4 - General transformational and sustainability issues

### General overview of the 100,000 Genomes Project

100,000 Genomes Project participants or carers had a positive view about the project and similar transformational projects (FG A, FG B, FG E). Other expert patients/members of patient groups, but non-participants of the 100,000 Genomes Project, were overall more sceptical (FG D, FG E).

### The consent process being embedded with the participants' disease pathway

Members said that currently the consent process of the 100,000 Genomes Project and their disease pathway are *'divorced'*, *'parallel but not linked'* (FG A). However, they could see links in the future, i.e. with diagnosis/treatment available and a card/records linked with the condition (FG A).

Some members felt it is viable to be embedded in the normal pathway for patients; it is for *'the greater good'* and it is the NHS' duty to adopt and adapt it (FG C). One member considered the project vital to reduce unnecessary intervention and improve treatment (FG C). Another thought it would have to be separate; expansion should be taken slowly to learn from findings (FG D).

Another felt that although it is a good project overall, it is disorganised and disjointed, with too much administration and not enough collaboration between the different units (FG D). Extra

reassurance should be offered that anxieties and limitations around the consent process and consent material would be eliminated (FG B, FG D).

Participants of one group suggested recruitment should become an established part of the cancer pathway, an option at the *'first given opportunity'* e.g. along with biopsy sampling. This may be supported with integration of the project into existing cancer or rare disease databases (FG B).

Concerns about the logistics of the project were discussed in another group, i.e. continuity after the pilot, availability of long-term staff and healthcare teams, and provision of feedback information in the long-term (FG E). Communication and coordination with GPs was also suggested, i.e. Information Technology linkage between specialists and GPs (FG E). Members questioned the project's impact on patients' care pathways in practice, and provision of appropriate feedback and support to members, including emotional support (FG E). This would depend on professional competency and capacity; *'as long as things are clear at each step of the project and it works smoothly'*, people would have trust and confidence. Appropriate support and information about its provision, e.g. peer support groups, coffee mornings, should be available to members (FG E).

### **The future of such projects**

Most members could see the benefits of such projects (all FGs). Members generally thought this kind of research should be embedded for the sake of scientific progress, will kick start new initiatives and will help the NHS and patients (FG D). Some recognised such projects as *'the way forward'* (FG B), as exciting, with a wider impact, and extra benefits (FG D). All members of one group recognised the importance of such projects to explore new knowledge and answer questions; there is a *'huge educational requirement about the possibilities of genetic testing'* and *'we must plough ahead with genetics health research, can't know how data will be used'* (FG E). One member could see a contribution to family and future generations of those with rare and genetic diseases and highlighted their unique position, how their experience makes them *'grateful'* for the project and more likely to oblige in the information and consent process (FG C).

Potential drawbacks included data insecurity, as *'genomic information may not be very secure in the future, and may be widely used and shared'*; blaming people for health states, preventing people having children, denying insurance etc. (FG E). Ethical implications and who makes decisions on access to and use of information, i.e. if NHS services become more privatised, were also discussed (FG E). Other members mentioned the high costs and the risk of creating an expensive field of personalised medicine (FG B, FG E), and the provision of appropriate emotional and psychological support (FG E).

Members felt that whether or not such projects have a viable future as part of routine care in the NHS depends on whether they show clear benefits, confidence in people's data being secure, confidence and trust in new projects and technologies (FG C). Their future is highly dependent on the NHS and *'whether it remains a united national, centrally controlled entity that is not privatised and remains accessible to all'*. Any future developments should be done with caution, and with regular public and transparent reporting on progress throughout the project's life (FG C).

Figure 9 Transformational and sustainability issues

*'If they were to create a system where they gave you a card, for example, or something even with a barcode on or a link to something, so that you could go to your professional and say 'My mum has taken part in the 100,000 Genomes Project...Look there are the findings.'...So that they can use it and that would help my descendents and the greater good as well...'* **100,000 Genomes Project participant, patient with rare disease**

*'Just focusing on the rare diseases, when we left hospital with my daughter we were told that in 70% of cases you'd never find out why she was this way. I'd like to know what that percentage is now and will be in the future. So just the way it's going to get answers for people has to be a good thing...finance and the right government will needs to be there, and once you get a diagnosis you need the support from the nurses and doctors.'* **GMC PPI Group member, parent of child with rare disease, non-participant**

*'...We've got to plough ahead with genomics and what it can do for us and medicine in our future. That's the most important. You can't keep saying, 'Well the insurance company might get this', they might very well but I'm more concerned about my future health and that of my family, and other people's families...'* **GMC PPI Group member, patient with cancer, non-participant**

## Additional barriers and facilitators

### Introductory Leaflets – flow, complexity, consistency of scientific/genomic terminology

The three-fold leaflet and suggestions for improvement and accessibility was discussed extensively (FG D, FG E). The leaflet was thought to include redundant sentences and negative statements (FG D, FG E); emphasis was suggested being put on treatment and prevention (FG D).

Concerns about consistency in language and information provided were raised, i.e. three-fold leaflet and ten-page leaflet (FG A, FG C, FG E). The third category of invited participants with 'severe infections' referred to in the ten-page leaflet was questioned (FG C, FG E).

### Accessibility and other formats

Availability of material in other formats, i.e. audio (FG A, FG B) and visual (FG B) were discussed; there was a consensus in one group that there are many accessibility issues to these materials (FG D). They are not accessible for the majority of English speakers, non-English speakers, and people with learning disabilities or dementia; in addition, they don't have sections for interpreters or carers (FG A, FG D).

An 'Easy Read' two-page document with key points in plain English, available alongside the other material, on paper and online, creating two-levels of information, was suggested (FG A, FG B, FG D).

### Commercial organisations and for-profit companies

Concerns were raised in two groups about commercial and for-profit companies' involvement, i.e. questioning pharmaceutical companies' role, the Project's 'money making' aspect, private companies purchasing the data to develop products to be sold to the NHS, potential forcible disclosure of findings by insurance companies (FG C, FG D). A member suggested clarification of the term 'insurance provider' on the PIS to specify health insurance or company insurance (FG C). The difference in a patient consenting from an 'altruistic' position as opposed to making money for the NHS or external companies was also highlighted (FG C). In addition, it was considered unfair for 100,000 Genomes Project participants to pay to access their own data (FG D).

### **Lack of publicity and awareness**

Members overwhelmingly identified the lack of publicity as an on-going barrier (FG B, FG C, FG E); one disliked the idea of *'Joe public spending money and no one knows what's going on'* (FG C). This may affect participation: *'participants will be going in cold'* (FG E).

They suggested TV and advertisements in magazines (FG B), GPs as well as hospital clinics (FG E) with more leaflets being provided directly by clinics (FG B); and the possibility to consent online (FG A). The use of social media, i.e. Facebook and twitter, was suggested to reach younger generations (FG A).

### **Data security (privacy) and data sharing (private companies)** – Including impact on insurance

Data protection and data sharing were discussed briefly in three focus groups (FG A, FG C, FG D).

Members were concerned about data security, potential entry points and overseas transfer (FG A, FG D). Other concerns were breaching confidentiality (FG B), and the language of data sharing, i.e. anonymised or pseudo-anonymised (FG A), anonymised or de-identified data (FG D). Who may misuse the data, the influence of genomics companies, information sharing with GPs, and the impact of the results on care were also queried (FG B, FG D, FG E).

### Summary

- 10 out of 13 GMCs were involved in arranging 5 regional focus groups (FGs) which were attended by 34 patient representatives or members of the public associated with the GMCs.
- There was agreement between the FGs that the consent documentation was long and difficult for members to understand; suggestions included more pictures, diagrams, and a glossary.
- There was support from some FG members to keep the majority of the information, all of which was seen as important in order to make a decision to participate or not.
- FG members provided detailed specific feedback on the PIS, consent forms and tri-fold leaflet with clear editing, format, layout, and style suggestions, and with the addition of a glossary.
- More information was requested by FG members about provision of information and access on emotional support and communicating with family.
- The FG members supported the current 100,000 Genomes Project option for participants to opt-in or opt-out of receiving additional findings results.
- The FG members had many suggestions on how to approach the transformation of the 100,000 Genomes Project into part of the routine NHS patient pathway.
- Some FG members wished for more publicity information to promote the benefits of the project.

## 3.2 Health Professional Recruiter Survey

The response rate for the survey was 36% (58/161). The denominator was calculated by asking the key contact within each GMC how many health professionals were active during the snap-shot period.

There were some differences between responses from recruiters for rare disease and for cancer. Therefore results are presented as responses by rare disease (RD) recruiters first followed by cancer recruiters second. **The cancer results are highlighted in bold.**

Nearly all GMCs (92.3%, 12/13) participated. The response rate varied between centres, ranging from 21 to 100% (median = 29%). The active number of health professionals per centre ranged between 3 and 29 (median = 4). Recruiting groups included; Nurses 36.2% (21/58), Doctors 22.4% (13/58), Genetic Counsellors 8.6% (5/58), and the 'Other' category i.e. those who did not identify with one of these groupings above, 32.8% (19/58). 13 of these 'Other' respondents stated their job titles, which were wide ranging and included; clinical genomic practitioner, clinical trials assistant, programme manager, project manager, project co-ordinator, research assistant, research associate, research technician. Almost a third of recruiters (32.8%) did not self-identify themselves as a nurse, doctor or genetic counsellor. Clinical specialties were wide ranging. The main group was genetics/genomics but others were; oncology, urology, cardiac, neuro-genetic, paediatrics and perioperative adult nursing (missing 22).

Two participants (ID numbers 55 & 71) were then excluded from the remaining data analysis because they did not complete the remainder of the survey questions, resulting in a data-set consisting of 56 participants (N=56).

Twelve recruiters, 21.4% (12/56), only recruited cancer participants, and 40 recruiters, 71.4% (40/56), only recruited rare disease participants. Four recruiters recruited participants for both arms of the project, 7.1% (4/56). Therefore we had N = 60 question set responses, 73.3% (44/60) rare disease responses and 26.7% (16/60) cancer responses.

**Q1. Which GMC are you part of? Q2. Please state your Health Professional Group.  
Q3. What is your clinical speciality? Q4. How many health professionals recruit in your GMC?  
Q5. In which section of the project have you recruited participants?**

- Q6, Q7** – Relate to the 100,000 Genomes Project participant information sheet (PIS)
- Q8** – Relates to specific open text feedback on a variety of consent materials
- Q9** – Relates to the 100,000 Genomes Project consent form
- Q10-Q16** – Relate to an 8 week recruitment period February and March 2016
- Q18** – Relates to support for recruiting staff

## Rare Disease and Cancer Recruiters

The results are presented as responses by rare disease (RD) recruiters first followed by cancer recruiters second. The cancer results are highlighted in bold.

**Q6. We would be interested in your views about the participant information sheet in its ability to help the participant make an informed choice about taking part in the project (n = RD44, C16)**

This question sought feedback on the participant information sheet in three specific areas – length, scientific terms and words, and amount of information. There was general support from the rare disease recruiters that the material was *'about right'* in terms of scientific terms and words (78.1%, 32/41) and the amount of information (58.5%, 24/41) it contained. However the majority (53.7%) thought it was overall *'too long'*. Also 22% (9/41) felt it was *'too complicated'* in terms of scientific terms and words, with 39% (16/41) stating they felt the amount of information was *'too much'*. Nearly all cancer recruiters felt was *'too long'* (**92.3%, 12/13**), with a similar proportion to the rare disease recruiters saying it was *'about right'* for scientific terms and words (**76.9%, 10/13**), and far more cancer recruiters felt it contained *'too much'* information (**92.3%, 12/13**).

**Q7a. Do you as a health professional providing consent think any further changes to the rare disease participant information sheet are required in the following three specific areas? (These specific areas have been highlighted in previous feedback) (n = RD44, C16).**

Recruiters felt that some changes were needed, as reflected by the response of *'maybe'* or *'yes'* in relation to flow of sections (45%, 18/40: **38.5%, 5/13**), consistency of use of scientific terms and words (41.5%, 17/41: **30.8%, 4/13**), and consistency of use of project terminology (52.5%, 21/40: **77%, 10/13**).

**Q7b. Please use this box to write any suggestions on ways to improve the participant information sheet in these specific areas.**

Summary data for all the open text questions is provided at the end of this section. The submitted comments have been provided to Genomics England to help inform the re-development of the consent materials.

**Q8. We are interested in your suggestions on ways we could improve any element of the rare disease consent materials \* select from a drop down list of all information sheets and forms.**

The submitted comments have been provided to Genomics England to help inform the re-development of the consent materials.

**Q9. We are interested in your perception of how often your participants have experienced difficulty in understanding each section of the participant consent form (n = RD44, C16)**

The following sections of the consent form were reported by more than 50% of recruiters to have caused participants difficulty *'sometimes', 'often' or 'very often'*.

Taking part (**53.9%, 7/13**)

Samples (50%, 20/40: **69.2%, 9/13**)

Access to my data and confidentiality (77.5%, 31/40: **76.9%, 10/13**)

Results (72.5%, 29/40: **76.9%, 10/13**)

**Q10, 11, 12, 13 and 15 relate to active recruitment during an 8 week period (February and March 2016).**

**Q10. During February and March how many participants have you recruited? i.e. been the health professional who countersigned their consent form? (RD40, C16)**

Of the 52 recruiters active during February and March, 36 completed the rare disease question set only, 12 completed the cancer question set only, and 4 completed both question sets, giving a total of 56 possible responses (n = 56, RD, 40 C, 16). Recruiters were asked to provide a best estimate for numbers of participants recruited during an 8 week period around February and March 2016. They were not asked to keep a log of recruitment so response is based on recall.

There was wide variation in the numbers of people recruited by each health professional. 753 rare disease and 98 cancer participants were reported to have been recruited by 48 respondents (8 responses missing). Respondents mainly reported recruiting *'10 patients or less'* (40%, 16/40: **69.2%, 9/13**). Approximately a quarter of respondents (25%, 10/40: **23.1%, 3/13**) recruited *'between 11 and 25'* and the remainder (35%, 14/40: **7.7%, 1/13**) *'26 and over'* (response n = RD40:C16).

**Q11. Considering these participants. Please indicate how many face to face or telephone appointments they had with YOU prior to signing the consent form.**

Q11 (n = RD 753, C98)	Qr11a		Qc11a		Qr11b		Qc11b		Qr11c		Qc11c	
Totals n = 851	1 Appt.		1 Appt.		2 Appt.		2 Appt.		3 Appt.		3 Appt.	
	n	%	n	%	n	%	n	%	n	%	n	%
<b>Participants consented</b>	676	89.8	70	71.4	63	8.3	18	18.4	14	1.9	10	10.2

\* The dataset contained one rare disease recruiter who reported consenting 110 participants all who took one appointment. When removed adjusted percentages are 88% (1 appt), 9.8\* (2 appt) and 2.2% (3 appt). (n = 643)

The vast majority, 89.8% (676/753) of rare disease and **71.4% (70/98)** of cancer consents took place during one appointment. However 10.2% (77/753) of rare disease and **28.6% (38/98)** of cancer consents took two or more appointments.

### Q12. Considering these participants. Please indicate the total time you spent in face to face or telephone discussion with the participant.

Totals	Q12a		Q12a		Q12b		Q12b		Q12c		Q12c		Q12d		Q12d	
RD 813 <sup>+</sup>	<30 mins		<30 mins		30min-1h		30min-1h		1-2h		1-2h		>2 h		>2 h	
RD 703 *																
C113																
	n	%	n	%	n	%	n	%	n	%	n	%	n	%	n	%
Pt's	246	30.3	51	45.1	435	53.5	50	44.2	122	15	12	10.6	10	1.2	0	0
	136	19.3*			435	61.9*			122	17.4*			10	1.4*		

+ Total participants consented increases here from the number stated in Q11 due to some recruiters completing Q12 who had not completed Q11.

\* The data-set contained one rare disease recruiter who reported consenting 110 participants all <30 mins. When removed this does affect the proportions and the adjusted percentages are presented in the table above and text below and are noted by \* (n = 703)

Recruiters reported that between a half and two-thirds of rare disease and cancer participants took '30 minutes to one hour' (61.9%\* 435/703), **44.2% (50/113)** to consent. About a quarter of rare disease participants took 'less than 30 minutes' (\*19.4% 136/703), compared to **45.1%** for cancer participants. There was evidence of longer consent times; 18.8%\* of rare disease participants took greater than one hour, and **10.6%** for cancer participants.

### Q13. Considering these participants. What proportion of these appointments took place during the participant's routine care clinical appointment? (n = RD44, C16)

Consent occurred during routine clinical care	Less than 25%		26-49%		50-74%		75-100%		75-100%		missing		total							
	n	%	n	%	n	%	n	%	n	%	n	%	n	%						
Respondents	29	78.4	0	0	3	8.1	0	0	1	2.7	1	7.7	4	10.8	12	92.3	3	3	40	16

The most common scenario for rare disease participants, occurring three-quarters of the time (78.4%, 29/37), was that 'less than 25%' of the consent contacts occurred within the participant's routine clinical care appointment. In contrast nearly all cancer participants (**92.3%, 12/13**) were 'consented during normal clinical care'.

**Q14. Within the last 2 months how often have your participants presented with specific needs which you needed to address as the consenting health professional during the consent process? (n = RD44, C16)**

From an option list of seven possible specific need areas highlighted by previous research, 50% or more of recruiters reported that for rare disease and **cancer participants (in bold)** the following areas '*sometimes*', '*often*' or '*very often*' caused difficulties for participants:

- 'Extra explanation needed to understand the participant information sheet & consent form' (85.4%) **(100%)**
- 'Low levels of literacy' (50%) **(33.3%)**
- 'Visual impairment' (35.9%) **(50%)**
- 'Increased levels of anxiety related to current health situation' (56.4%) **(100%)**

Issues are similar to those stated by rare disease recruiters, but cancer recruiters suggest specific needs relating to participants' anxiety related to their current health situation, and less need relating to low levels of literacy.

**Q15. During February and March, approximately how many patients have you met who declined participation? (n = RD40, C16)**

58.3% (21/36) of rare disease recruiters stated that '*no-one had declined the project*' (4 missing). This is in contrast to **16.7% (2/12)** cancer recruiters who said that '*no-one had declined the project*' (4 missing). A further 38.9% (14/36) of rare disease recruiters had '*between 1 and 10 decliners*', the remaining (2.8%) had '*20 to 31 decliners*' and had recruited 30 in total. A rare disease recruiter who consented 110 participants reported '*no decliners*'. The majority of cancer recruiters, **83.3% (10/12)**, stated that '*between 1 and 10*' had declined (4 missing).

**Q16. What factors might have influenced these participants to decline?**

Summary data for all the open text questions is provided at the end of this section. The submitted comments have been provided to Genomics England to help inform the re-development of the consent materials.

**Q17. How confident do you feel in delivering a quality consent process to potential participants of the 100,000 Genomes Project? (RD44, C16)**

Of the rare disease recruiters, 25% (10/40) felt '*very confident*', 62.5% (25/40) felt '*confident*', and 12.5% (5/40) felt '*somewhat confident*'. None selected '*not confident*' or '*prefer not to say*' (4 missing). Of the cancer recruiters, **16.7% (2/12)** felt '*very confident*', **66.7% (8/12)** felt '*confident*', and **16.7% (2/12)** felt '*somewhat confident*' (4 missing).

**Q18. Are there any ways in which Genomics England could support you as a health professional more with the delivery of a quality consent process?**

Summary data for all the open text questions is provided at the end of this section. The submitted comments have been provided to Genomics England to help inform the re-development of the consent materials.

**Open text summary – Rare Disease Recruiters (Questions rQ7, rQ8, rQ16, rQ18).**

There were alternative perspectives present; comments were received for the materials both to be more concise and more detailed (sometimes asked for by the same recruiters).

- Where materials could be more concise
  - Where individual points made are ‘wordy’ or could be represented by bullet points
  - Where it is repetitious e.g. between key points in ‘blue box’ and long text
  - Where it seems superfluous e.g. text on Mental Capacity Act
- Where more information may be needed, on particular areas that patients are concerned about
  - What are the possible ‘additional findings’?
  - On specific additional tests
  - On ‘carrier status’
  - When main findings can be expected
- Applicability to particular patient needs
  - Patients physically unable to sign a consent form
  - Those with poor educational levels/literacy (suggestion is to use the 11-15 booklet)
  - Preparedness to read a long document and really engage with it “people want to be told where to sign!”
  - Those better suited to images/audio/HCP reading information to them (produce audio?)
  - Children (who might not consider themselves ‘ill’)
- Physical/online materials
  - Large number of items to initial (one signature on each page is enough?)
  - Consent form too long and repeats PIS
  - Too many bits of paper
  - IT can be very clunky (e.g. the EDCT, uploading consent materials)

**Family dynamics engaging with materials**

- Partners of patients may feel left out as they can’t be ‘participants’, especially if they want to have children
- More emphasis needed on where are ethical concerns regarding other people/family; can staff support this? Sometimes people decline as they worry about impact on family
- Sometimes the patient wants to take part but other family members do not; siblings refuse having blood taken

**Consent and Participation Process**

- Lack of time (needing to leave at certain time), clinics too busy to embed recruitment
- Participants often need to come back just for recruitment (HCPs try to combine these with clinical review)

- Comes at a bad time when people cannot cope with thinking about another complex issue
- Can create anxiety in some
- Need for a blood test (esp. those are afraid of needles/children)
- Some people just lack understanding/interest
- Helpful to contact patient in advance and gauge interest

#### **Particular concerns with the project for patients**

- Data security who owns data/data storage
- Related issues of insurance, reluctance especially for commercial companies to have data

#### **What would be helpful to the consenting process**

- Additional information/different formats
  - Information on audio
  - Information on DVD
  - Images to support the information
  - Shorter booklet with more additional materials
- Changes to the materials as outlined above e.g. simpler, shorter, fewer 'bits of paper', use of images and video
- Promotional material to display in clinics; wider publicity about the project
- More training, in particular
  - Briefing on additional genetic conditions that may be found/on carrier status
  - Top-up training as it makes a lot more sense having had the experience
  - Having done the genetics quiz should be mandatory as part of consent training
  - Face-to-face training sessions
- Less targets and pressure especially for small teams
- Some argued for nominated, specially trained staff to do the recruiting

#### **Responses from cancer recruiters. Comments were very similar to the rare disease recruiters' responses but a few were unique to patients with cancer:**

- Issues around explaining 'reproductive findings'
- Huge amount of information received after diagnosis of cancer, or weeks before a major operation, this comes at a bad time
- Sometimes patients are at the clinic just for surgery
- Need for a short genomic counselling course focusing on feelings/reactions
- Possibility to consent to having a tumour sample stored and rethink later?

## **Summary**

In summary, the health professional survey covered Genomic Medicine Centres with a varied number of recruiters from a variety of professional backgrounds. A limitation is that based on the estimated numbers of recruiters who were sent the survey, our response rate is 36% (58/161). It is possible that some recruiters did not know about or receive the email survey link. Two reminders were sent by the local contact (usually the programme lead) and reassurances obtained that all health professionals had been given the opportunity to respond. It is possible that those recruiters who chose not to respond had differing demographics or views from those presented here. This low

response rate, although comparable with on-line surveys of NHS staff, means that responder bias has to be considered when inferring outcomes from the survey data.

Many of the health professionals who responded provided detailed suggestions on how Genomics England could change the current consent process or materials and these have been provided to Genomics England to help with the development of the consent materials. Some health professionals stated the wish for further training and on-going development and had suggestions on how this could be achieved.

- 58 recruiters out of the 161 who were active during the period responded, providing their experiences of recruiting participants (response rate 36.5%).
- The additional consent materials were welcomed (especially the introduction leaflet) and were seen as useful in informing participants. Recruiters asked for more promotional materials to raise awareness.
- Recruiters commented that the consent form was too long and repeated information in the participant information sheet; and questions were raised as to why multiple signatures were required and not just one.
- Many suggestions were made as to how the materials/documentation could be improved e.g. consistency of terminology, less dense, more pictures. Suggested changes to the materials e.g. simpler, shorter, fewer 'bits of paper', use of images and video.
- The importance of the health professional recruiter in the consent process was stressed with the written material needing verbal explanation of genetic concepts and clarification during the majority of consent interviews.
- 50% or more recruiters stated that they perceived participants experienced difficulty in understanding the following sections of the consent form; 'taking part', 'samples', 'access to data and confidentiality', and 'results'.
- There was concern from the recruiters about the length of time taken to consent (69.7% of rare disease patients took *'more than 30 minutes'*, in contrast to 54.9% of cancer participants) and also the timing of the discussion especially for anxious newly diagnosed cancer patients.
- Rare disease recruitment seemed to be occurring in special clinics. 78.4% of rare disease participants were recruited *'not during their routine care appointment'* in contrast to 92.3% of cancer participants who were recruited *'during their routine care appointment'*.
- 87.5% of rare disease recruiters and 83.4% of cancer recruiters were *'confident'* or *'very confident'* in delivering a quality consent process.
- Suggestions were made to Genomics England regarding how they could support the recruiting staff to deliver the consent process e.g. training updates (face to face), information on timescale for results, training on additional findings, carrier status, patient emotional response, improve IT project infrastructure, advice on how to respond to patient queries (once in the project), introducing dedicated recruitment staff as clinical staff may not have the time.

### 3.3 Participant Survey

#### Overview

12 out of 13 GMCs participated in the survey. Responses per GMC ranged from 2 to 49. As there is large variation in 100,000 Genomes Project recruitment between GMC sites this is not unexpected.

No individual GMC contributed more than 26% of the returns, thereby the results represent a range of GMC activity during the snapshot 8 week period.

174 responses were received, 87.8% (153/174) rare disease and 12.2% (21/174) cancer from a total of 1196 participants recruited (1045 rare disease and 151 cancer), giving a response rate for rare disease participants 14.6% (153/1045) and cancer participants of 13.9% (21/151), and an overall response rate for the participant survey of 14.6%. The denominator for response rate is based on Genomics England weekly reporting figures for the 8 week period when cards were being handed out.

Two rare disease postcards were completed by children and their results are not included in the main analysis as research ethics permissions did not cover seeking children's views. Notably, the open text comment on the card was – “why no option for children's views?”

Comparing the responses given by the cancer participants with the rare disease participants there is unlikely to be any statistically significant differences between the rare disease and cancer participant response groups. Therefore the results below are presented together for the two groups.

## Rare Disease & Cancer (n = 172)

### Demographics

Nine participants responded using the individual password controlled on-line option and 163 by returning a response postcard. Forty percent of respondents were male (64/160) and 60% female (96/160), 12 missing. The majority 75.5% (129/171) were between the ages of 25-64, with 2.9% (5/171) between the ages of 16 and 24, and 21.6% (37/171) 65 or over, 1 missing. Responses were received mainly from affected adult patients with a rare disease 31% (52/198) and from the unaffected parents of a participating child 31% (52/168) and also from adult patients with cancer 11.3% (19/168), 4 missing. All participation categories were represented.

### Q1. Please tick which of the following materials you read or viewed prior to consenting to participation in the 100,000 Genomes Project.

The majority of 100,000 Genomes Project participants had seen or viewed the Participant (92.5%) or Consultee (32.2%) Information Sheet & consent form. The Introductory Booklet was read or viewed by 36%. However the videos and animations (5.6%) and flip chart (7.5%) had not been read or viewed by many participants, 11 missing responses for this question.

### Q2. How do you rate the .....

Q2	Poor		Satisfactory		Good		Excellent		missing	total
	n	%	n	%	n	%	n	%		
..overall satisfaction with your consent experience	0	0	3	1.7	35	20.3	134	77.9	0	172
..written information given about the project	1	0.6	9	5.3	51	30.2	108	63.9	3	172
..verbal information given about the project	1	0.6	1	0.6	35	21.0	130	77.8	5	172
..care and attention you have received	0	0	0	0	27	16.1	141	83.9	4	172

The majority 98.3% (169/172) of participants rated their overall satisfaction with the consent experience as 'good' or 'excellent'. They rated elements of the consent experience as 'good' or 'excellent' for written 94.1% (159/169), verbal 98.8% (165/167) and overall care 100% (168/168). They were least satisfied with the written information.

Chi-square analysis was performed to detect if overall satisfaction rating was associated with the following variables; type of disease, patient category, gender or age group. No statistically significant associations were found apart from that older age groups are more likely to read the PIS in more detail than younger age groups ( $p < 0.02$ ), and men are more likely to rate satisfaction with verbal information highly than women ( $p < 0.02$ ).

### Q3. How much of the participant information or consultee information sheet did you read?

The majority, 66.1% (113/171) 'read all the sheet in detail'. However 32.2% (55/171) 'scanned but did not read in detail', and 1.8% (3/171) 'did not read any', 1 missing (n = 172).

### Q4. What did you think about the participant or consultee information sheet you read?

Participants were asked to feedback on three areas. There was overwhelming support that the PIS was 'about right' as regards length (88.7% 149/168), scientific terms and words 96.4% (163/169), and amount of information (92.9%, 157/169). 10.1% (17/168) felt it was 'too long' and 4.7% 8/169 felt it contained 'too much' information.

### Q5. In the information sheet would you have liked more or less detail to be included about?

Q5	Less		Right		More		Missing	Total
	n	%	n	%	n	%		
<b>Use and storage of samples</b>	6	3.6	151	90.4	10	6.0	5	172
<b>Results related to main condition</b>	5	3.1	121	74.2	37	22.7	9	172
<b>Additional results unrelated to main condition</b>	7	4.3	130	79.8	26	16.0	9	172

Approximately three-quarters of participants felt that the amount of information included in the PIS was 'right'. However 22.7% (37/163) of participants would like 'more information' about results related to their main condition and 16% (26/163) 'more information' about additional results.

### Q6. Did you feel you were given enough time to understand all the information prior to signing the consent form?

Nearly all participants, 99.4% (165/166), answered 'yes' to this question.

### Q7. Would you have been satisfied with a shortened information sheet as the more complete text is available on line?

All participants answered this question and views were divided. 26.2% (45/172) would have been satisfied 'with a shorter information sheet if the complete text was on-line', 36.6% (63/172) would not. There were 36.6% (63/172) who were undecided and gave a 'maybe' response.

**Q8. Were there any bits (document or process) of the consent experience that were difficult or confusing?**

This was the only opportunity for participants to write feedback on the postcard. The area for response was however limited to one line only, which may have deterred longer responses. There were 113 comments, 91.1% (103/113) consisted of 'no', 'nope', 'none'. The others are presented below. n = 113

<b>Text</b>	<b>n</b>
<b>Deciding whether to ask for carrier information to be given to me</b>	1
<b>I particularly appreciated the lady .....explaining to my young daughter about extracting DNA and blood cells</b>	1
<b>Poor wording in some areas which caused confusion</b>	1
<b>Some</b>	2
<b>Some issues about consent are quite confusing</b>	1
<b>Too much information to read through, needs to be quicker and easier to read, appreciate it is complex project</b>	1
<b>Unfortunately process takes very long time</b>	1
<b>Was a bit long worded, could be more concise</b>	1
<b>Yes, the question regarding "do you want to know about other results...if the project discovers I might be at higher risk of developing another disease" it's a very big Q</b>	1

**Q12. Are there any ways we could improve the consent experience? (Only asked on the on-line option to complete the survey)**

The following suggestions/comments were made.

<b>A film showing more information showing what happens with your samples</b>
<b>Having a CD/DVD to watch may have helped.</b>
<b>I would like to have seen a film showing the process of what happens from the time your blood is taken</b>
<b>No/None (2)</b>
<b>Whole process &amp; discussion were clear &amp; concise. Many thanks!!</b>

## Summary

The overall response rate for the participant survey was 14.6% (174/1196). We received assurances from the key contacts at each GMC of the date when the postcards were available to be handed out to potential participants.

Response rates can be influenced by many things such as survey distribution, perceived benefits, and demographics (Aldridge Alan 2001). The NHS England Friends and Family test is similar in design to the feedback postcard used in this study. Estimates show that between 18-34% of eligible patients complete the Friends and Family survey (NHS England 2014). This shows that the response rate for the participants in this study is below what might be expected.

Response bias also needs to be carefully considered when interpreting the results of the participant survey. It is important not to over-generalise the results considering that they are provided by only 14.6% of those who were consented for the 100,000 Genomes Project during the snap-shot 2 month period. For instance, measures of satisfaction have a common-sense and political appeal, but they are the measures that experts, including experts in quality improvement, consider the least useful on their own for improving patient experiences locally (Mazor KM, Clauser BE et al. 2002, The Health Foundation 2013). Some key findings are outlined below but should be used with caution as participants who did not complete the survey may hold differing views.

174 out of the 1,196 participants (of the 100,000 Genomes Project) who consented during the period completed feedback postcards:

- 100% of participants rated their satisfaction with the care and attention they received as *'good'* or *'excellent'*.
- 98.3% of participants rated their overall satisfaction with their consent experience as *'good'* or *'excellent'*.
- 94.1% of participants rated their satisfaction with the written materials as *'good'* or *'excellent'*.
- However 32.2% *'only scanned and did not read the PIS in detail'*, and 1.8% *'did not read it at all'*.
- The vast majority 88.7-96.4% felt the length (89%), scientific terms (96%) and amount of information (93%) in the participant information sheet was *'about right'*. The main criticism being that 10.1% felt it was *'too long'*. This was in contrast with health professional recruiters where 92.3% thought it was *'too long'*.

## 4 Synthesis of Activities

### Synthesis methods

Up to this point the results from the three stakeholder groups had been analysed independently of each other adhering to the measures of quality and validity appropriate for methodology used (Qualitative and Quantitative). This chapter reports the Synthesis stage of the analysis, following the parallel mixed designs framework (Teddlie 2009), the final stage is a meta-inference of the results from all three activities. The following iterative process was undertaken.

- During a project team meeting the data from each of the three activities was discussed and similarities and differences between stakeholder groups were noted. Evidence from each stakeholder group was collated across the 10 original pre-defined areas of focus.
- The evidence was then re-read and grouped into facilitators (things which stakeholders reported were going well with the consent process) and barriers (things they thought could be improved). It was collated under the original pre-determined 10 key areas of focus for the evaluation. When examining the best presentation for the evidence obtained, the original 10 pre-determined areas of focus were collapsed down to five and then presented as four synthesis tables
- A synthesis table was then produced for each of the four areas of focus and included suggestions made by each stakeholder group are presented as a third column in the tables.

- The Simplemind visual network app. was used to view the data and to assist with this process where evidence was tracked back to the stakeholder group using icons.

**Focus Groups = People**



**Health Professional Recruiters = Mortar Board**



**Participants = Rosette**



- The tables and the Simplemind diagrams were then iteratively reviewed and draft recommendations made for presentation to Genomics England. The evidence supporting the recommendations is tracked back to the underpinning evidence by cross-reference to the synthesis tables (recommendation numbers placed alongside evidence in the synthesis tables).

## Visual Mapping

Figure 10 Example of visual mapping which helped create the synthesis tables



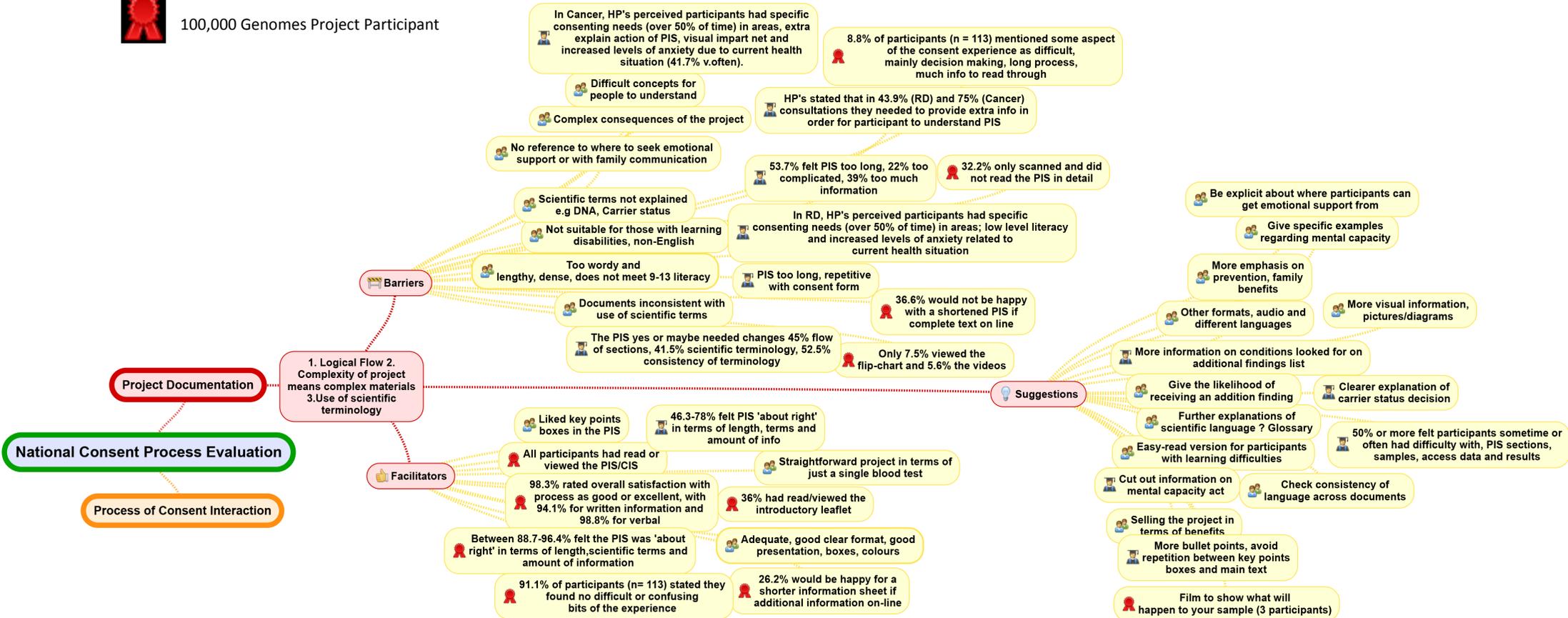
Focus Group Comment



Health Professional Recruiter Comment



100,000 Genomes Project Participant



## Synthesis Tables

Table 1 Pre-determined areas of focus (1, 2, 3) – Logical flow (1), complexity of consent materials (2), use of scientific terminology (3)

Facilitators (things going well)	Barriers (things to consider changing)	Suggestions to consider
<p>(FG) There was support for the recent changes and additions (video/information leaflet) and for the key point boxes, colours and formatting. (1,3, 5)</p> <p>(HP) About half felt the RD PIS for RD was 'about right' in terms of length, scientific terms and amount of information. (3)</p> <p>(P) The vast majority of participants (88-96%) felt the PIS was 'about right' in terms of length, scientific terms and amount of information. (1,2,3)</p> <p>(P) 94% rated the written materials as 'good' or 'excellent'. (1,2,3)</p> <p>(P) 91% participants (n = 113) stated they found no bits of the consent experience difficult or confusing. (1,2,8)</p>	<p>(FG) The majority opinion was that they would not suggest reducing information but that currently the PIS and Consent Form are still 'too wordy, dense, do not meet 9-13 year old literacy levels and there are gaps and inconsistencies in use of scientific terminology. (3,9,11,12,14)</p> <p>(FG) Some members did not feel the PIS 'sold the project well' in terms of personal patient and family benefits (RD) and value to the NHS in terms of prevention and treatment of disease in the future. (20)</p> <p>(FG) The focus groups were critical of the three-fold DL leaflet 'The 100,000 Genomes Project' as it was unclear who the audience for this was and many terms not explained. (16)</p> <p>(HP) 53.7% felt the RD PIS was 'too long', 22% 'too complicated and 39% 'contained too much information'.</p> <p>(HP) 92.3% recruiting cancer participants felt PIS was 'too long' and contained 'too much information'. (9,11,12,13,21)</p> <p>(HP) 50% or more of the participants they recruited had had 'sometimes' 'often' or 'very often' had difficulty with consent form sections – <i>samples, access to data and results</i> sections. (6,8,14,15, ,17,,18)</p> <p>(P) 32% of participants only scanned but did not read in detail the PIS.(9,12,17,22)</p> <p>(P) Only 7.5% viewed the flip chart and 5.6% the videos/animations. (13)</p>	<p>Aim to increase the number of participants who actually read and engage with the PIS;</p> <ul style="list-style-type: none"> <li>• having a 'glossary of terms' document</li> <li>• having more visual images/pictures/diagrams in PIS</li> <li>• A new 'easy read' version or the option of being able to use the 'young persons' leaflet for adults if low levels of literacy</li> <li>• An 'easy-read' version for participants with low levels of literacy, learning difficulties</li> <li>• re-write to meet 9-13 literacy standards</li> <li>• reduce repetition between PIS and consent form and between key information boxes and text</li> <li>• add section on emotional concerns and where emotional support (psycho/social and family communication) may be obtained from</li> <li>• Non-English and low vision versions</li> <li>• shorter PIS for cancer participants</li> <li>• Consider why the need to include seemingly lengthy text on Mental Capacity Act</li> <li>• Produce an audio version of the PIS, more suited for people who do not process text information well</li> <li>• To include the name of next of kin/family member to whom results to be given on cancer consent form</li> <li>• Re-design of the one-page tri-fold leaflet if intended for patient use.</li> </ul>

Table 2 Pre-determined areas of focus (4, 7) - Issues related to main and additional findings, including opt in/opt out (4) and seeking broad vs. specific consent in relation to additional findings (7)

Facilitators (things going well)	Barriers (things to consider changing)	Suggestions to consider
<p>(FG) Strong preference to keep the option of testing for additional findings and the ability to opt-in and opt-out (dynamic consent). <b>(7)</b></p> <p>(P) 74% of participants felt the information in the PIS on main condition and 80% on additional findings was ‘about right’. <b>(1,2,3)</b></p>	<p>(FG) However there was uncertainty about where participants would go for further information about the conditions currently tested for, support in making the opt-in/out-out decision and guidance on how to talk to family members about topics. <b>(15,17,,18)</b></p> <p>(FG) A list and more information, i.e. links, available to participants on the conditions tested for now and updated as testing changes in the future. <b>(15,13,18)</b></p> <p>(FG) A realistic indication to participants when main finding results might be expected. <b>(15,13,18)</b></p> <p>(HP) 33% of RD HP and 23% of cancer HP recruiters stated that participants ‘often’ or ‘very often’ had difficulty with the results section of the consent form. <b>(15,17,18)</b></p> <p>(HP) Some HP stated that patient confusion or concern regarding results (both main findings and additional findings) were the reason why some decided to decline participation. <b>(15,17,18)</b></p> <p>(HP) Some HP stated that patient concern over possible or unknown impact of the results on family members had been a reason for patients to decline participation.<b>(17,18)</b></p> <p>(P) 23% of participants would like more information on the PIS about the main condition. <b>(15,18)</b></p> <p>(P) 16% would like more information on the PIS on additional findings. <b>(15,18)</b></p>	<ul style="list-style-type: none"> <li>• Signposting participants as to where they may get an opt-in/opt-out form</li> <li>• Provision of a pre-paid return envelope where they could send back opt-in opt-out forms after their initial decision</li> <li>• Patient accessible on-line list of currently looked for and reported additional findings on the Genomics England website with links to trusted patient focused information sources.</li> <li>• More information on the currently tested for conditions and implications for family members.</li> <li>• More health professional training on how to explain these conditions, especially carrier conditions or where to signpost patients for more information.</li> <li>• More training for health professionals in describing what the opt-in choice will mean for participants</li> <li>• Information available as to when realistically participants are likely to receive results related to main findings or additional findings</li> <li>• To provide a number/estimate of the chance that a participant might receive an additional finding</li> <li>• More information in the PIS about additional findings and process of receiving main results</li> </ul>

Table 3 Pre-determined areas of focus (5) Data security, privacy and sharing (5)

Facilitators (things going well)	Barriers (things to consider changing)	Suggestions to consider
<p>(P) 90% of participants felt that the amount of information in the PIS on use and storage of samples was 'about right'.</p>	<p>(FG) There was lack of clarity of who gains access to data and at what time-points. The data-sharing language is unclear i.e. anonymised vs. pseudo-anonymised, de-identified. <b>(14)</b></p> <p>(FG) Some members raised concern about disclosure of personal information to insurance companies. <b>(13,14,)</b></p> <p>(FG) Some members had concerns regarding the influence private genomic companies has on the NHS. <b>(13,14)</b></p> <p>(FG) Some felt there needed to be greater clarity in the participant materials that there is a money making element of this, with private companies accessing data – for the benefit of the NHS. <b>(13,14)</b></p> <p>(HP) Recruiters stated that two thirds of participants 'sometimes', 'often' or 'very often' experienced difficulty with the RD and Cancer consent form section 'access to my data and confidentiality'. <b>(9,11,13,14)</b></p> <p>(HP) Some stated that patients may have declined participation due to concerns related to data security, ownership and reluctance for commercial companies to have the data. <b>(13,14)</b></p>	<ul style="list-style-type: none"> <li>• Clarify the data-flow, who has what access to what portion of data and when e.g. specialist, GP, NHS England, researcher, private company</li> <li>• Be clear at what points it is considered anonymised or pseudo-anonymised, de-identified, and use examples of 'real life' scenarios/requests to explain access rights</li> <li>• Consider a video showing 'what happens to my sample and data after I donate it'</li> <li>• Consider additional training and materials to support the health professional recruiters in this area</li> </ul>

Table 4 Pre –determined areas of focus (6, 8, 9, 10, 11) - Nature and timing of approach (6), time taken to consent (8), consent interview (9), flexibility and pre-determined pathway (10) and transformation/legacy of project (11)

Facilitators (things going well)	Barriers (things to consider changing)	Suggestions to consider
<p>(FG) Members who were participants have had good experiences of the consent process. Describing various approaches in operation face to face in clinic, by email, by telephone, open day or a combination. <b>(1,3,5,8)</b></p> <p>(FG) Members felt it was important to give the HP adequate time to consent families and additional time for those needing extra support (e.g. learning disabilities, new cancer diagnosis). <b>(8,18,23)</b></p> <p>(HP) 90% of rare disease consents took place at one appointment vs. 71% for cancer participants. The rare disease appointments nearly all took place OUTSIDE of routine clinical care, whereby three-quarters of cancer consent were WITHIN their routine care appointments. <b>(18,22,23)</b></p> <p>(HP) 54% RD and 45% of cancer consents took less than 30 minutes. <b>(18,23)</b></p> <p>(HP) There were a diverse range of consenting professionals with 36% nurses, 22% doctors, 9% genetic counsellors and 33% others (who did not identify with groups above), these consisted of clinical trials assistants, research assistants and research technicians. <b>(19,20)</b></p> <p>(HP) Number of participants consented per recruiter during an 8 week period varied</p>	<p>(FG) Some members were critical about the consent process; too much administration and disorganisation and not enough collaboration. <b>(18,19,20)</b></p> <p>(FG) There was concern raised about NHS England’s ability to provide appropriate feedback and support to participants. <b>(15,22)</b></p> <p>(FG) there were concerns about the logistics of the project, i.e. continuity after the pilot, availability of long-term staff were discussed in another group. The ability of healthcare teams to pick up that work after the project ends and feedback of information was questioned by participants. <b>(8,17,22)</b></p> <p>(HP) 70% of RD and 55% of cancer consenting took over 30 minutes to complete. <b>(8,17,22)</b></p> <p>(HP) 16% of RD consents and 11% of Cancer consents took over 1 hour. <b>(8,17,21,22)</b></p> <div data-bbox="640 1018 1207 1129" style="border: 1px solid black; padding: 5px; margin-top: 20px;"> <p>This diversity could also prove a barrier for adequate training and updating.</p> </div>	<ul style="list-style-type: none"> <li>• Consider mechanisms to support the time required for participants to learn about the project, assimilate the information and make an informed decision to participate.</li> <li>• Importance of interaction and support from HPs.</li> <li>• Provide patients with information about social and psychological support organisations/groups</li> <li>• All participant materials available on-line and easily found by patients</li> <li>• Balance between meeting recruitment targets and providing time for consent process</li> <li>• Clarity in what support mechanisms will be in place for families already in the project when recruitment closes</li> <li>• Given the diverse range and backgrounds of people taking consent some assessment of capability of consent might be considered – formal assessment, suggestions included making a pass level for the genetics quiz or GCP training mandatory for consenting staff</li> <li>• The usual pathway for rare disease consenting seems that the patient is invited for a ‘consent taking’ appointment outside of routine clinical care and this differs from the cancer participants – Perhaps need to consider differing support and consent pathways for the two groups</li> <li>• Participants overwhelmingly were very satisfied with the process overall and only a few suggested areas which had been difficult or confusing (limitation on postcard methodology)</li> </ul>

Facilitators (things going well)	Barriers (things to consider changing)	Suggestions to consider
<p>between 1 and 110 (rare diseases) between 1 and 25. <b>(19,20)</b>            (HP) 38% GMCs had 1 to 5 recruiters active, 26% between 6 and 10, and 36% from GMCs with over 10 <b>(13)</b></p> <p>(HP) 87% of rare disease and 83% of cancer recruiters felt 'very confident' or 'confident' in delivering a quality consent process. <b>(8)</b></p> <p>(P) Overall 100% of participants rated the <i>care and attention they had received</i> during the consent process as good or excellent. Cancer participants were less likely to rate excellent than RD participants (70% vs.85%). <b>(8,18,19,23)</b></p> <p>(P) Overall 99% of participants rated verbal information about the project as good or excellent. <b>(1,8,18)</b></p> <p>(P) 98% of participants rated their overall satisfaction with the consent experience as good or excellent. <b>(1,2,3,8,18)</b></p> <p>(P) Of 113 responses to the open question – any bits found confusing or difficult – 91.1% reported 'none' and often added very good comments about the role of the HP in the process. <b>(1,2,3,8,18,19)</b></p> <p>The participants were equally split between whether they would want a shortened PIS if more information was available on-line – 26% 'yes', 37%, maybe, 37% 'no'. <b>(4)</b></p>	<p>(HP) Comments – clinics too busy to embed recruitment, running out of time, often difficult to combine with routine clinical care, some suggested nominated specially funded staff for this project. <b>(17,18,,21,22)</b></p> <p>(HP) The need to have a blood test vs. saliva and also having to re-schedule to come back is a cause for declining participation. <b>(21,22)</b></p> <p>(HP) Cancer – recruitment comes at a very difficult time for the patient as they have just been told they likely have cancer, very emotional and difficult to consent at this time without overburdening the participant. <b>(21,22)</b></p> <p>(HP) 13% of rare disease and 17% of cancer recruiters only felt 'somewhat confident' in their ability to provide a quality consent process. <b>(18,19)</b></p> <p>(P) Of the 8.9% of open text responses to 'were there any difficult elements of the process' – these included process takes a long time, decision making regarding additional findings takes a while, too much written information. <b>(15,17,18)</b></p>	<ul style="list-style-type: none"> <li>• Health professionals were involved in summarising and translating/explaining the context of the project, definitions and scientific concepts to patients – this takes time.</li> <li>• Health professionals suggested having more promotional materials so that they could raise the profile of the project locally</li> <li>• Focus groups felt integration of the project into routine NHS care needed much careful thought and it would need robust funding and sustainability planning.</li> <li>• Focus groups would like to see regular reporting on progress, which is made public and transparent, throughout the project's life, to inform the transformability aspects.</li> </ul> <p><u>Health Professional identified training needs</u></p> <ul style="list-style-type: none"> <li>• Education on conditions on additional findings list</li> <li>• Short genomics course on feelings/reactions/communicating genetic information with family</li> <li>• Education on carrier testing</li> <li>• Support how to discuss with patients how to involve their family members</li> <li>• Top-up training</li> <li>• Provision of list of where families can go for psycho/social support for their condition.</li> </ul>

## Synthesis summary

Four descriptive categories (meta-inferences) were developed from the synthesis activity. The summary needs to be read taking account of the methodological restrictions and strengths of this evaluation. The strengths lie in the fact that all GMCs contributed in some way and that views were elicited from three stakeholder groups, representing the geographical spread of the project. Also the evaluation was focused on an 8 week period where the consent material or processes did not change and where reliable estimates of response rate could be obtained.

**1. “Differing views”** - This category describes the finding that there were differing views provided across this evaluation. This is to be expected as participants in this evaluation will have differing perspectives and experiences which will influence their responses. Of interest were the similarities between the health professional recruiters and the non-participant focus group members, who tended to be most critical of the consent materials and process. 100,000 Genomes Project participants who attended the focus groups and those who completed postcards were overwhelmingly supportive of the project and the care they had received.

**2. “Dialogue, Decisions and Dependency”** - This category describes the important factors highlighted by many respondents in that deciding to take part or not in the 100,000 Genomes Project required dialogue between many people - potential participants, their families, the routine clinical care team and the consenting staff of the 100,000 Genomes Project. There were many examples of different pathways leading to the patient declining or agreeing to participation. Throughout this category the major issue of importance was the clarity of consent materials in informing and documenting discussions and decisions. The front-line recruiting staff are dependent on their GMC for the infrastructure and management as well as to the central 100,000 Genomes Project team for central advice and materials. Recruiting staff felt confident in providing a quality consent process but also identified areas where they would welcome additional training and requested an on-going supportive infra-structure for protected time to take consent and also updating.

**3. “Support & Follow up care”** - Most stakeholder groups highlighted the importance of providing patients/participants with more information regarding support especially with emotional and practical issues arising due to being a part of the 100,000 Genomes Project. Areas highlighted were ‘family communication’ and ‘psychological support’. Issues were mentioned about the need for clarity regarding; what happens with data and results after a participant had consented, how to manage participants’ expectations and feedback regarding how samples are to be used by researchers and private companies. Of importance was clarity for participants regarding the pathway by which information (result found or not) will be fed back to them and the timescale for this.

**4. “Improvements”** - This category describes the finding that both the recruiters and the focus group members felt that the PIS and consent form for both Rare Disease and Cancer participants should be improved. Many helpful suggestions were made. The responses from 100,000 Genomes Project participants were very satisfied with the consent process and their care and they felt no significant changes should be made – but this finding may not be generalisable across all participants due to the low response rate (14.6%). Also under this category are improvements which were suggested to the timing of the consent discussion, especially for the cancer participants, where scheduling an extra 30 minute consent discussion at a time when the patient is anxious about diagnosis and treatment is proving difficult. Also recruiters commented on the burdensomeness of the Information Technology systems for recording consent and sample collection.

## 5 Recommendations

This snap-shot evaluation (mid-way through the 100,000 Genomes Project) showed that there were differing opinions both within and between stakeholder groups. This means that when considering the development of materials to help facilitate consent, it is important to bear in mind that how they are perceived and utilised will differ in relation to context and the individual participant's information needs.

The results of this evaluation need to be considered alongside on-going and published research as well as ongoing policy debate regarding consent for whole genome sequencing.

There was support for the supplementary materials developed to help with the consent process, such as the information leaflet, animations and flip-chart. These were viewed as being useful in preparing patients for the consent discussion. Suggestions were provided concerning how to support NHS recruiting staff, including on-going training. Overall, staff who completed this survey (who represented 36% of active recruiting staff at the time of the evaluation) felt 'confident' or 'very confident' in being able to provide a quality consent experience. Although not necessarily representative of all participants, due to the response rate of 14.6%, those 100,000 Genome Project participants who returned a feedback postcard (n = 172) were extremely satisfied with the consent materials and process they experienced. Participants in the focus groups were also supportive of the information included in the consent material. There were aspects of the consent materials and consent process that received positive comment from all stakeholder groups.

However, evidence of where the materials could be improved was also found. Concern was raised by recruiters and focus group members regarding the length and readability of the participant information sheets (PIS) and consent forms and they proposed specific changes that could be made. These are detailed in the 'National Service Evaluation Project: Background Document' which is available to Genomics England for review when implementing changes to the consent material or process. Half of the recruiters perceived that participants 'sometimes', 'often' or 'very often' experienced difficulty with certain sections of the consent forms, especially the sections '*taking part*', '*samples*', '*access to my data and confidentiality*', and '*results*'. Approximately half of the recruiters reported that participants 'often' or 'very often' needed '*extra explanation to understand the project*', had specific needs related to '*low levels of literacy*' and '*increased levels of anxiety related to current health situation*'. All stakeholder groups felt it was important to keep most of the current informational content in some way within the materials but suggested changes to the PIS and consent documents in terms of presentation and readability.

**The following recommendations are made to Genomics England and are based on the evidence from this evaluation. It is expected that Genomics England will take forward the conversations with partners across the healthcare system such as NHS England, Genomic Medicine Centres and Health Education England.**

Recommendations 1 to 8 are suggestions on elements to maintain, recommendations 9 to 17 are issues Genomics England may consider changing in relation to project documentation, and recommendations 18 to 26 are linked to project processes. Each recommendation is linked back to the data collected by providing the recommendation number alongside the evidence within the synthesis tables in Chapter 4.

#### **Things to maintain (1-8)**

1. The current range of consent materials i.e. participant information sheet (PIS), consent forms, information leaflet, animation/flip chart.
2. The current wording in the PIS relating to explanations of scientific and genomic terminology in line with recommendation 9.
3. The majority of the information content within the PIS (responses from participants and most focus group members support this, along with 50% of recruiters).
4. The PIS both in paper form and on-line (not just on-line).
5. The key point boxes and colour on the forms.
6. Continue to develop accessibility to consent materials for low vision patients.
7. The ability of participants to opt-in and opt-out of the search for additional findings.
8. The facility for health professionals to have the time to adequately discuss and answer patient questions prior to the consent decision.

#### **Changes to Consider - Project Documentation/Accessibility (9-16)**

9. Improve the presentation and readability of the PIS and consent forms in the following ways.
  - 9.1 Reduce the density of words.
  - 9.2 Reduce replication of text especially the sections between the consent form and PIS and also after the key point boxes.
  - 9.3 Use pictures/diagrams where possible.
  - 9.4 Work with participants/100,000 Genomes Project National Participation Panel/PPI groups/Ethics Advisory Committee to review new material/changes to assist in approaching standards such as the plain English guidelines and the Accessible Information Act.
  - 9.5 Reduce the length of the consent form.
  - 9.6 Review the terminology (e.g. main findings, additional findings) used across all project materials, including written and animation/video to assist with understanding the project specific terms.
  - 9.7 Include a source for standard definitions/concepts (this could also be available for recruiters) for example, a glossary of definitions of both scientific and project terminology and ensure these are consistent with descriptions in the text.
10. Continue to support non-English speakers by promoting the use of existing NHS translation services when discussing project documentation with potential participants.
11. Produce an 'Easy Read' version of the PIS and consent form that could be used for participants with reduced intellectual ability or those for whom English is a second language (possibly the 'young adults' PIS could be adapted for use with this group).

12. Provide an audio/video of the content of the PIS and consent form for participants who find access to the written word difficult.
13. Promote the Genomics England website as a place to access information (presented in a variety of formats) about taking part in the project.
14. All materials to adopt consistent use of terms relating to anonymisation, when is it linked-data, when unlinked and clarify at which point this occurs e.g. at what point companies might have access.
15. More information to be available to participants regarding the concept of additional findings and especially ones of reproductive significance, including the timescale for results and future contact.
16. To clarify the audience for the tri-fold information booklet and if it is for use with patients, review the booklet and add definitions of terminology used.

### **Changes to Consider - Process of Consent Interaction (17-22)**

17. Explore ways of ensuring that recruiters have appropriate time to discuss the issues raised in the PIS with participants, where this is needed.
18. Support recruiters through on-going consent training including how to manage questions from participants regarding support with communication in their families and social/psychological support.
19. Support recruiters in the information technology systems for registering participants and logging samples.
20. Provision of more publicity materials to help recruitment into the project.
21. A staged consent process for cancer participants if anxiety levels are high at the time of diagnosis. Possibly consider adapting the PIS and consent forms for this patient group.
22. In the further integration of whole genome sequencing into routine NHS clinical care, the following issues need careful consideration and planning.
  - 22.1 Maintaining adequate time for consent discussion.
  - 22.2 Consider carefully the changes required for the transition of Whole Genome Sequencing consent from the research environment into routine clinical care.
  - 22.3 Supportive appropriate local mechanisms for receipt of main findings and additional findings (results).
  - 22.4 Support and updating of staff providing patient support.
  - 22.5 Consideration of consent timing within the routine care pathway (especially cancer patients and their families).

## Acknowledgements

The research project team would like to thank all the 100,000 Genomes Project participants who completed postcards, and the 34 members who attended focus groups across the country. Also to the health professionals who completed the survey. All of whom have provided detailed feedback which we hope will inform the development of the next phase of consent materials, as well as inform the wider debate concerning consenting for whole genome sequencing.

The research team would also like to thank the members of NHS England and Genomics England who have offered invaluable support in communications with the GMCs and with hosting the survey on the Genomics England website, also with the initial co-design of the evaluation. All GMC PPI leads or other associates who helped organising the focus groups. Thanks to the Steering Group who have been involved with the planning stages and development of the Schedule and the drafting of the final report.

This project was financially supported by Genomics England, The Innovation Agency (Academic Health Science Network North West Coast, University of Central Lancashire (UCLan), University of Birmingham and Imperial College London.

## Glossary

### Consent documentation

Any material produced by Genomics England to provide potential participants with information about participation in the 100,000 Genomes Project (including all REC approved materials) and also the consent forms used for the participant to provide evidence of consent.

### The consent discussion process

This includes how potential participants (existing NHS patients and their family members) became aware of the 100,000 Genomes Project, their interaction with health professionals with whom they might discuss the project, and also health professionals who are involved with facilitating the discussion and documenting informed consent.

## References

1. Aldridge Alan, L. K. (2001). Surveying the social world - principles and practice in survey research. Buckingham · Philadelphia, Open University Press.
2. Braun, V. and V. Clarke (2006). "Using thematic analysis in psychology." Qualitative Research in Psychology **3**: 77-101.
3. Creswell, J. Piano Clarke, V L (2011). Designing and conducting mixed methods research. Los Angeles, CA,, Sage.
4. Department of Health. (2007). "The Mental Health Act 2007." from [http://www.legislation.gov.uk/ukpga/2007/12/pdfs/ukpga\\_20070012\\_en.pdf](http://www.legislation.gov.uk/ukpga/2007/12/pdfs/ukpga_20070012_en.pdf) Accessed Nov 2016.
5. Dozois, E., M. Langlois, N. Blanchet-Cohen and J.W. McConnell Family Foundation (2010). A practitioners guide to developmental evaluation. [www.mcconnellfoundation.ca/de/resources/publication/de-201-a-practitioner-s-guide-to-developmental-evaluation](http://www.mcconnellfoundation.ca/de/resources/publication/de-201-a-practitioner-s-guide-to-developmental-evaluation). Accessed November 2016.
6. Genomics England website [www.genomicsengland.co.uk](http://www.genomicsengland.co.uk). Accessed November 2016.
7. Greene, J. (2007). Mixing methods in social enquiry. San Fransisco, Jossey-Bass.
8. IBM Corp (2013). IBM SPSS Statistics for Windows, Version 22.0. Armonk, NY, IBM Corp.
9. Mazor KM, Clauser BE, Field T, Yood RA and G. JH. (2002). "A Demonstration of the Impact of Response Bias on the Results of Patient Satisfaction Surveys. Health Services Research. 2002;37(5):1403-1417. doi:10.1111/1475-6773.11194." Health Services Research **37**(5): 1403-1417.
10. NHS England. (2014). "NHS England Review of the Friends and Family Test." from <https://www.england.nhs.uk/wp-content/uploads/2014/07/fft-rev1.pdf>. Accessed November 2016.
11. NHS Improving Quality. (2015). "The NHS Change Model <http://www.nhs.uk/capacity-capability/change-model.aspx> " Accessed November 2016.
12. Teddlie, C. T., A (2009). Foundations of Mixed Methods Research: Integrating Quantitative And Qualitative Approaches In The Social And Behavioral Sciences. London, SAGE: 152.
13. Teddlie, C. T., A (2009). Foundations of Mixed Methods Research: Integrating Quantitative And Qualitative Approaches In The Social And Behavioral Sciences. London, SAGE: 266.
14. The Health Foundation. (2013). "Measuring Patient Experience." Evidence Scan, from <http://www.health.org.uk/sites/health/files/MeasuringPatientExperience.pdf> Accessed November 2016.