Public deliberation: research access to Newborn Genomes Programme data

A findings report

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Contents

Executive Summary

In this report we describe what participants involved in a public deliberation about research access to Genomics England's Newborn Genome Programme said. It highlights key considerations for Genomics England when thinking about:

- The scope of discovery research using newborn genome data.
- Research conducted in the context of specific scenarios.
- Reactions to data linkages in different health contexts.
- Communications and transparency around the programme.
- Considerations on trust and trustworthiness with genomic research.

1. Introduction

The aim of the deliberation was to develop a wide-ranging process through which public participants could explore whether there are unique features of the Newborn Genomes Programme that would warrant a different approach to enabling and communicating research access and use.

103 participants, drawn from a demographic broadly reflective of the population came together in four sub-groups:

- Northern and Southern England participants met online in four workshops between 7th and 23rd February 2023
- Participants from Liverpool, London and surrounding areas met in day-long workshops held in-person on 4th and 25th February 2023

The deliberative focus

The public deliberation focused on the second aim of the Newborn Genomes Programme: understanding how babies' genomic data could be used for discovery research, focusing on developing new treatments and diagnostics for NHS patients.

It was explained to participants that discovery research would take place within the National Genomic Research Library (NGRL). This is a secure database managed by Genomics England which contains genome and health data from thousands of individuals (many of whom have a rare disease or cancer) who provided consent.

Participants also heard that the Newborn Genomes Programme would use an 'all-in consent offer'. This means that parents will be asked to sign up to the use of their baby's genome and linkage to clinical data to allow the following:

- Return of actionable findings to newborn's family
- Research on newborn screening
- Research on broader healthcare questions (within the NGRL acceptable uses)
- Recontact to request follow up data related to newborn screening research or to offer opportunities to participate in other studies
- Use of any of the baby's leftover sample for further research.

Speakers from the Genomics England team, and researchers involved in discovery research, spoke to participants during the deliberative process. This gave them information on the programme and potential areas for research which informed their deliberations. Detail on the information given is provided in Appendix 1.

2. The findings

Two key points are worth highlighting at the beginning of this summary for their importance to the Newborn Genomes Programme.

1. Whilst participants frequently referred to the data within the programme as 'sensitive', this sensitivity is not referring to the use of newborns' data. Rather that this is genomic data and as such is perhaps the most sensitive data human beings can have. Participants believe that trust in the discovery research programme will be contingent on it being clear that the research respects the sensitive nature of this data.

2. Throughout the report we find that participants were often more concerned about the motivations behind a proposal for discovery research than what the purpose of the research might be. They felt that understanding why a researcher would want to conduct the research is important for the approvals process, particularly in ensuring that the research has a public benefit.

What we found when discussing research access for discovery research using data from the Newborn Genomes Programme is summarised based on the five findings chapters in this report.

Reactions to discovery research

Participants came to the deliberation with different levels of knowledge and experience of discovery research. Those with more experience tended to support discovery research within the programme. Those with less experience felt more concerned that discovery research is too exploratory and lacking in specifics to give confidence.

As participants worked through the process, many highlighted the benefits in discovery research that they could see. This focused initially on discovering new diseases and information about genetic conditions. As they heard more about the possible outcomes from discovery research, participants began to see particular benefits for pharmacogenomics. They thought that the concept of tailoring medicines to genomes sounded like a very exciting opportunity. Other benefits participants identified included discovery research that:

- Improves the life expectancy of people with specific conditions e.g. cystic fibrosis
- Provides opportunities for early intervention strategies and support for those with genetic conditions
- Provides a pathway to a more prevention-focused model for healthcare.

Participants were supportive of discovery research that might be considered higher risk (e.g. Finding out about being at risk for a condition) *if* it might lead to an outcome in the longer term that could benefit people with severe life limiting conditions.

Concerns about discovery research focused on:

- Risk of harm from a security lapse or data breach within the National Genomic Research Library (NGRL)
- Data manipulation to enhance a researcher's career, or in other ways skew

the results
- A fear of the unknown – inherent in any discovery research programme, where there might be unintended consequences from not having a clear expected outcome from the research
- Trepidation that the approvals process for research to access the data may not be robust enough to prevent those with motivations other than public good using the data in unethical or unauthorised ways
- Researcher motivations – where profit is not seen as an acceptable reason for conducting discovery research.

Participants called for existing governance structures to give greater clarity to researchers, to study participants, and to wider society on what is an acceptable use of the data, and how this is assured. They also wanted to know that there are systems in place if something goes wrong.

Consent was key to the dialogue discussions. Participants felt more confident in the scope of the discovery research if they were equally confident that the data is collected using robust and ethical consent processes. Parents, and the newborn as it grows up, must be assured that they can withdraw from the study at any time.

Discovery research scope and impact

Participants expected that discovery research using newborn genomic data will focus on identifying rare conditions and finding ways for earlier diagnosis and treatment. The impacts of the discovery research with real-world implications are important to participants. Such impacts identified by participants included:

- Saving money on discovery research by minimising diagnostic odysseys
- Showing where there are gaps in knowledge which need filling with further discovery research
- Enabling the UK to become a world leader in this area.

Words of caution from participants on the scope of discovery research included:

- Discriminating against people or communities who might have a greater susceptibility to particular genetic conditions, particularly when those people feel that their communities have not benefited, or have been harmed by discovery research in the past
- Increasing the life span of populations as a result of discovery research findings could prolong life at the expense of planetary resources

Although the focus of the deliberation was on the discover research aim, participants also brought in views on the screening aim of the study. For some this raised a concern that an increase in prenatal screening could lead to the eradication of certain traits in people. A particular cause for great concern is seen in society trying to screen out difference.

Mission creep is also seen as a cause for concern with some asking, ‘How far should this go?’. Going beyond the remit of individual research projects was also seen as a worry for some, e.g. if somehow their work within the NGRL becomes so interesting to them that they veer off the initial approved research study into a new area. This is a particular concern when this extension beyond the approved remit might have a profit motive, and/ or has not had ethical approval.

Research uses in relation to our scenarios

Five scenarios were discussed in the dialogue[1]. These were developed by staff from Genomics England with experience in genomic research. Each scenario enabled participants to explore a particular facet of discovery health research, including data linkages with non-genomic health and social data. The main points that emerged from these discussions are that:

- Linking data is broadly welcomed by participants provided it is proportionate to the research question and re-identification is avoided
- Some conditions, e.g. obesity, are complex and incorporate factors beyond genomics, such environmental and social factors. These factors were significant for participants, indicating that data linkage to non-genomic datasets could be important.
- Longitudinal research using linked datasets could be a powerful tool in better understanding conditions and the health of the nation.
- Greater understanding of rare conditions was seen as a highly significant purpose for discovery research.

Participants used the discussions on scenarios to prioritise different types of discovery research. Conditions that are life limiting or with severe ongoing impacts, such as Rett syndrome or epilepsy, were a high priority for discovery research for participants because finding treatments or earlier diagnosis will have significant benefits for those with these conditions and their families.

The full range of factors participants used for this prioritisation combined aspirations for the research and for the wider health and care ecosystem. The prioritised list of factors includes:

- Understanding severe, rare and life limiting genetic conditions, prioritising this as a reason for approving discovery research
- Understanding the symptoms of a condition and the potential for a cure
- Potential to lead to an earlier and more definitive diagnosis
- Potential for research leading to earlier health benefits, particularly in childhood
- Preventing misdiagnosis and unnecessary medication
- Potential to create personalised medicines
- Potential that research using newborns' data could inform research into other conditions affecting children and adults – advancing knowledge on a wider range of conditions than those being studied
- Research into conditions where there are no or limited existing treatments e.g. for rare conditions
- Research that can inform both medical (e.g. improved medications) and social interventions (e.g. improved specialist support in schools for children with ADHD) was seen as important
- Research that could lead to reductions in NHS and social care costs
- How rare or common the condition is – the balance between finding treatments and cures for a condition which affects many people, to supporting a very few people who might live with a very severe condition.

[1] Research into: epilepsy, obesity, ADHD, Rett syndrome, Condition X.

The severity of the condition is for many an important factor through which the need for discovery research should be assessed.

Further exploration of responses to linking datasets:

Participants were broadly positive about linking health datasets for discovery research. They were reassured when participants perceived that linking data sets for discovery research might lead to:

- An improved understanding of genetic conditions
- An improved understanding of environmental risk factors for certain conditions
- A more rounded picture of the data from this largely healthy cohort of newborns (rather than just having genomic data)

Participants highlighted specific instances of where dataset linkage might be particularly important:

- Establishing longitudinal studies where greater insight can be gained from looking at linked datasets over time
- A greater understanding of the conditions that disproportionately affect some people in society e.g. sickle cell disorders and their impact on people with Black African and Caribbean heritage.

Gaining a fully rounded picture of genetic conditions by linking datasets was seen as the real value of the process. Participants were concerned, however, when they considered the possibility (intentional or inadvertent) of re-identification.

Some participants stressed the 'power' of data when linked. Many felt that this could increase the potential for public benefit accruing from discovery research. Others, however, indicated that this 'power' could be used for harm, particularly when research findings are manipulated for political or financial gain.

Participants were asked specifically to react to the possibility of maternal health data being linked to newborn genome data. Again, participants were broadly positive about this option. For many it felt like a 'logical next step' which did not conflict with the aims of the Newborn Genomes Programme. Some participants struggled with this idea when they considered that there would be no direct benefit for the mother or child in this data linkage. Others were also concerned that this proposal might cause anxiety to the mother, particularly if she her health record includes information that she does not want to share e.g. about her lifestyle or abusive events in her history.

Communications and transparency

Consent is a key concept for participants. Whilst parents will be asked for consent during pregnancy, it remains a discussion point for participants when thinking about the long-term nature of discovery research. Participants called for clarity throughout the programme on what parents will be consenting to, how to withdraw consent, and what kinds of communications are likely to come from being part of the programme.

Participants are ambitious for Genomics England's communications strategies. They see a role for multimedia and broadcast media in raising awareness of a) the programme and b) discovery research. Particularly as an 'all-in' consent model is being used.

Re-contact of parents if a new condition, treatment or therapy is found

The question of whether and how to recontact parents if a new condition, treatment or therapy is found through discovery research was discussed by participants. For some, re-contacting parents in this scenario is essential, and part of Genomics England's duty of care to study participants. However, this is contingent on it being clear to parents of newborns during the consent process that this recontact is a possibility. Recontact should be done sensitively with decisions made with care on who makes the contact; and how that contact should be made. Having support in place for families is essential.

Communicating what discovery research is being done, who is involved in the research, and, longer-term, what the outcomes of the research has been were all seen as essential to making people aware of the value of the programme.

Considerations on trust

During the dialogue, participants' reflections on trust have led to a set of principles which characterise a trusted person, organisation or service. They are:

Act with **transparency**, using clear communications and with the expectation of openness in all relationships.

Define, and act within, high **ethical** and **moral standards**.

Be **reliable** and **proactive**: say what you are going to do, do it, and tell people that you have done it.

Be **genuine**: show that you care, you are empathetic, and you will stand up for what you believe in.

Be **discreet**, what I share with you shouldn't be shared with anyone else unless I've agreed to it.

Put **safety** first. Do nothing to harm people or knowingly put them at risk and have safeguards in place for when things do go wrong.

Demonstrate that **public benefit** comes before financial motivations.

Commit to a **mutually beneficial relationship** and be clear about what both sides are committing to.

Of these, transparency is the principle that comes to the fore as highly significant for trusted relationships.

Dialogue participant expectations of researchers who access the NGRL are that:

- They have been through a rigorous approvals process, which in the case of for-profit organisations should have additional stringent assessments of their motivations for conducting the research.
- They have a robust research plan which has a clear rationale for the need to research using genomic data
- Are committed to sharing their findings in ways which are accessible and are useful to families and wider society.

Participants throughout the dialogue referred to newborns' genomic data being 'sensitive'. **We found no evidence that the sensitivity they refer to is related to the fact that data will come from newborns**. Rather, sensitivities focused on the fact that the data is genomic and is integral to a person's make-up, questions of identity, and 'who I am' genetically. This mirrors the findings from the public dialogue: Implications of Whole Genome Sequencing for Newborn Screening (HVM/ Genomics England, 2021). Dialogue participants see genomic data as perhaps the most sensitive data of all and should be treated with enormous respect by all those involved in the both the main study and for discovery research. The trustworthiness of the programme is reliant on this being the case.

Fairness and an inclusive approach to data collection and identification of discovery research topics was seen as important, and should be applied to:

- Who is included in the main study
- Where they live
- The research that is undertaken.

Nothing should be done through the programme to reinforce bias or prejudice or remind people of historical discrimination in health research. For example, participants with experience of sickle cell disease felt that only when their voices were heard was the condition taken seriously, because their communities had not been listened to previously.

What Genomics England needs to do to demonstrate its trustworthiness

Given participants' focus on good communications and the high priority they place on transparency, it is clear that there a number of steps Genomics England can take to be recognised as a trustworthy guardian for this sensitive and valued data. These steps are rooted in the considerations on trust set out in above and are set out below.

1. Transparency is key to trustworthiness

Participants stressed that transparency is a prerequisite of the study's trustworthiness. They wanted Genomics England to communicate widely and consistently about the whole programme and its discovery research aspects. The ways of doing this that participants explored are set out in Chapter 5, but include:

- Annual updates on what discovery research is being done with the data
- Reporting back on research findings
- Sharing success stories
- Explaining when things go wrong, or don't turn out as expected

2. Raise awareness

Many participants said they were unaware of Genomics England and previous projects such as the 100,000 Genomes Project before taking part in the dialogue. They felt it would be hard to demonstrate that Genomics England is a trustworthy organisation without first raising awareness of the organisation and its work.

3. Promote Genomics England's relationship with the NHS

Some participants felt that Genomics England could demonstrate its trustworthiness if its relationship with NHS is spelled out. They felt, particularly since the pandemic, that the NHS is highly trusted across society and if it were well known that NHS clinicians and other staff are involved in the programme it would reassure people that Genomics England can be trusted.

A few participants went further, and said that an efficient way of demonstrating trustworthiness would be for Genomics England to move under and NHS umbrella.

4. Show your credentials

Just as participants want to ensure researchers have the right skills, experience and credentials to conduct discovery research with genomic data, equally it is important that Genomics England staff are shown to be experts. Such expertise would include demonstrating that they understand the field, know how to set up the data management processes and can set up robust approvals/ and ethics review processes. Participants advised that clearly stating on Genomics England's website and in other communications who the staff are, and their track history, is essential.

5. Highlight the safeguards in place

Being very clear about the safeguards in place for the principle of putting safety first is an essential demonstration of trustworthiness. When discussing this, participants emphasised the importance of:

- High levels of data security
- Informed consent, with a clear process for withdrawing at any point
- Deidentification of the data – with explanations of what 'deidentification' means clearly expressed to all those who join the study
- Establishing and communicating what the process is if something goes wrong
- Showing a track record in successfully managing the NGRL and previous projects which have involved researchers being granted access to sensitive genomic data.

6. Be accountable

Participants took Genomics England's role in designing, delivering, and managing the Newborn Genomes Programme very seriously. As such they wanted to know that Genomics England will act responsibly and take responsibility for the decisions made now and in the future about the study. This includes the establishment of an oversight committee with diverse members bringing lived and professional experience to ensure Genomics England stays true to its commitments. If Genomics England demonstrates that they are accountable for what happens in the programme, it will be trusted.

1. Introduction

1.1 Background

About Genomics England

Genomics England works with the NHS to bring forward the use of genomic healthcare and research to help people live longer, healthier lives. Genomics is a ground-breaking area of medicine that uses our unique genetic code to help diagnose, treat and prevent illnesses. Established in 2013, Genomics England launched the world-leading 100,000 Genomes Project with the NHS, demonstrating how genomic insights can help doctors across the NHS, and building a foundation for the future by assembling a unique dataset.

Genomics England is working with patients, doctors and scientists to improve genomic testing in the NHS and help researchers access the health data and technology they need to make new medical discoveries and create more effective, targeted medicines for everybody.

About Hopkins Van Mil

Hopkins Van Mil is a dynamic and successful independent social research agency. We create safe, impartial and productive spaces in which to explore and gain an understanding of people's views on the content which matters to them, to stakeholders, and to society. We work flexibly to build trust. HVM has extensive experience in preparing for, designing and facilitating effective deliberative processes. We hold a lens up to issues which are contentious, emotionally engaging and on which there are a broad range of viewpoints that need to be taken in to account.

About the Newborn Genomes Programme

The Newborn Genomes Programme is an NHS-embedded research study that will explore the benefits, challenges, and practicalities of sequencing and analysing the genomes of newborns through using whole genome sequencing (WGS). The evidence gathered from the study will be evaluated rigorously to inform decisions about whether to roll-out this technology and develop the world's first national newborn screening programme that uses whole genome sequencing.

Aim two of the Programme's three objectives (figure 1) is to understand how newborns' genomic and health data could be used for discovery research – for example, to enable the development of new diagnostics and treatments. With parents' consent, babies' genomes could be de-identified and added, alongside their health data, to the National Genomic Research Library (NGRL). This would enable carefully vetted academic, clinical, and life science industry researchers to develop novel diagnostics and treatments as well as understand how current therapies can be improved or repurposed, offering better outcomes to NHS patients.

The public deliberation focused on this second aim. The aim of the deliberation was to develop a wide-ranging process through which public participants could explore whether there are unique features of the Newborn Genomes Programme that would warrant a different approach to enabling research access and use.

Figure 1: The three aims of the Newborn Genomes Programme

Discussions focused on participant views on the acceptable and appropriate scope of research using this data, and how trustworthiness can be demonstrated with respect to data access.

The deliberation was commissioned in January 2023; the fieldwork took place in February; and the coding, analysis and reporting in March 2023.

1.2 Who took part

103 people took part in the deliberation. We recruited from across England drawing from urban, rural and suburban communities. The group was a broadly reflective sample which was weighted to increase, in relation to current census data, the number of people drawn from communities experiencing racial inequalities, disabled people, those with long-term health conditions, and people with and parents of children with genetic conditions.

1.3 The dialogue process

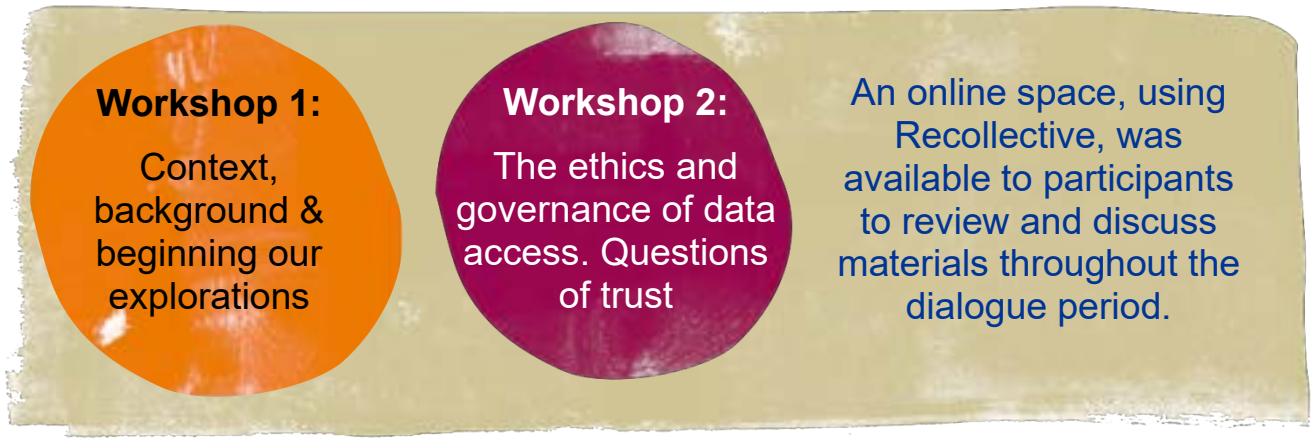


Figure 1: The dialogue process

The dialogue took the form of two full day workshops (on 4th and 25th February) for participants in London and Liverpool who took part in-person. For those who took

part online (groups from Northern and Southern England) the workshops took place over four week-day evenings from 7th to 23rd February.

1.4 A note about this report

This report explains what we heard from those who participated in the dialogue. Readers of the report with an interest in the methodology and process can find full details at Appendix A.

We have used qualitative research methods to review what participants told us. Transcripts were created from each of the consultation methods used. These were anonymised so that no one can be traced back to the comments that are included in this report.

Qualitative research reports, including this one, do not report on the number of times something was said, but rather the strength of feeling expressed across the methods used. For this project we used grounded theory, which means we read, and re-read, the transcripts many times. We collated what was said into key themes and used those themes to draw out meaning from the discussions. We chose this approach to ensure the findings are rooted in what participants said, rather than looking for confirmation of preconceived ideas. Throughout the report:

- Bullet points are used to summarise key points made. These mostly reflect areas of agreement and where points were made by many people across many of the groups
- Terms such as ‘a few’, ‘several’, ‘some’ or ‘many’ are used to reflect particular areas of agreement and difference

Anonymised quotations are used to highlight points made by a number of participants and to underline points made by a range of people. They also highlight points of particular significance to participants. These quotations are not edited so as not to distort the speaker’s meaning.

2. Reactions to discovery research

We begin with participants' initial reactions to discovery research. On joining the dialogue, participants had a range of levels of knowledge about discovery research. Those participants who knew more at the beginning tended to support discovery research because they felt that research leads to greater knowledge, even if the results are inconclusive or negative. Those who knew less tended to feel that discovery research is like 'looking for a needle in a haystack' or 'browsing'. In general, participants were not familiar with genomic research specifically and at times struggled to understand how researchers could use genomic data to identify new conditions.

> *"They're not saying they already have an idea of what they're looking for, they're just, kind of, going in blind."* Participant, Liverpool in-person group

2.1 Benefits of, and hopes for, discovery research

Participants were introduced to the concept of discovery research through a series of tasks on a dedicated online community space (Recollective) available to participants throughout the dialogue, and presentations given during the workshops by the lead facilitator and external specialists (see Appendix A). To further explore this concept, they were asked in their small discussion groups to brainstorm potential discovery research that they could think of that researchers might want to undertake using National Genome Research Library (NGRL) data.

In general, uses identified by participants at this stage focused on the discovery of new diseases.

> *"So, we thought as they research into, say, perhaps known genetic conditions, it might help the discovery of ones which are unknown."* Participant, London in-person

Participants were struck by the opportunities that discovery research presented towards developing personalised medicine. This was a particular feature of one specialist presentation. Many participants thought the use of discovery research to develop tailored medications was a very positive use of the data.

Throughout the dialogue, participants often explored the opportunities for genomic research in relation to their own experiences of health conditions. This example is no different in that their response to personalised medicine relates back to their own experiences, or those of people they knew, in the treatment and responses to medications for a range of health conditions.

> *"With personalised genome profile, over time there will be less and less trial and error for patients, and more genetically tailored and, therefore likely to work, medications. Obviously it is a complex and*

lengthy process to map DNA and find the markers and correlations, but that's why the research is so important, there's a lot to be discovered" Participant, Recollective

"I find the [genetic factor in] drug reactions interesting as I have been hospitalised for a week with a reaction to medication since our last session" Participant, Recollective

Participants also related strongly and positively to hearing examples of how discovery research has helped to improve outcomes for specific conditions, such as improving life expectancy for people with cystic fibrosis. They heard about this in the London in-person workshops from an expert speaker, and also through an online clip on Recollective. Examples which include specific conditions tended to resonate more positively with participants than general ideas about discovery research. The more hypothetical the example, the more unknown and potentially frightening to participants.

"[Watching the] cystic fibrosis explanation made it clearer for me to understand why wanting to know about genomes was interesting and informative" Participant, London in-person group, Recollective

*"So when researchers talk about research in very general terms [...] I tend to think of issues/concerns I have with exploratory research and how it might be used, while the CF video makes me realise how important and life-changing such research can be in leading to positive health outcomes and improved quality of life for many people now and in the future."*Participant, Recollective

In addition, participants identified the benefits of discovery research as providing opportunities for more preventative and early intervention strategies. They thought this could help to prevent conditions deteriorating, put support in place, and encourage lifestyle changes to support the management of a condition. Some participants believed this indicated a future with a more prevention-focused model for healthcare, which they saw as resulting in health system savings.

"Early identification of genetic differences for just earlier treatment. And the example I used for that was a TV show that I watched, like a few weeks ago. About a family who had a mitochondrial disease, and in one of the children specifically, they couldn't figure out what it was. Because there was no other

indication of where the symptoms were coming from, other than the genetics. So it would save a lot of invasive treatments and diagnostic stuff." Participant, Liverpool in-person group

"I was just wondering potential cost saving as well, if they've been genetically determined to develop a disease from a much earlier point as well, would that be liable to save costs in a way they could have head straight away to a specialist instead of going through processes and waiting for it to develop?" Participant, Liverpool in-person group

There is an interesting parallel to be seen between the 'trial and error' nature of discovery research and its ability to prevent 'trial and error' in diagnosis and treatment for patients (especially children). This means that participants in general were positive towards scientists and researchers undertaking exploratory and discovery research with a higher level of risk e.g. not leading to a positive outcome, because they felt that this could help to make more precise treatment available to those with certain conditions.

"They might find that there is a link between obesity and genetics or there isn't. Or, you know, genetics is part of it and then environmental factor is-, then they can go on to do a wider study that says what are the other parts of the factors in it. But if they don't go in and explore they don't know. So, for me personally I think every data access request that we've talked about I think is, sort of, fair game." Participant, Southern online group

"There's a lot of trial and error. I mean they've got experts that work on that, and neurologists, but it must be very, very concerning for a parent who's got a child who could develop multiple seizures a day, to try and find ways to help treat that. But, I mean if this could lead to treatments for those children, then, yes, I would say it's a good idea for them to have access to that data to look into this." Participant, Northern online group

2.2 Worries and concerns about discovery research

Whilst some participants expressed concerns directly about discovery research

taking place, the majority of concerns were about the possibility of negative outcomes occurring as a result of what discovery research relies on, i.e. large quantities of genomic data stored in the NGRL.

Some participants were concerned about the motives of those doing discovery research. These participants felt that scientists and researchers could not be trusted not to manipulate the data to fit a desired result, in the interest of their own reputation. They therefore did not trust the results of the research.

> *"We know scientists make things look the way they want it to look and I'm not being, you know, negative of it but you just don't want to go down that route of making the data match our hypothesis because that's what we want it to be."* Participant, Online Southern group

Amongst some participants there were indications of fears of the unknown when it came to discovery research. This means that alongside support for finding new diseases, there was also trepidation. Without specific examples, some participants thought there are likely to be as-yet-unknown risks and consequences, which made them feel worried about discovery research.

> *"What makes me feel a bit uncomfortable is what other negatives that could come of it? The knowing too much."* Participant, Northern online group

How data is securely stored, accessed, and managed

A common worry amongst participants focused on the possibility of data breaches affecting the genomic data held as part of the Newborn Genome Programme for discovery research purposes. Whilst many participants were reassured about the information security processes which are currently in place in the NGRL, other participants remained concerned that there could be a breach of this secure environment. Despite being told that the NGRL is a Trusted Research Environment (TRE) and a 'reading' not a 'lending' library, some participants remained concerned that researchers might be able to 'copy' the data and take it out of the secure environment. Some participants were particularly concerned about people in other countries being able to access the data, where they felt the governments of those countries did not operate ethically. Specific mentions were made of China and Russia in this context.

> Participant 1: *"They also said that the data is so large that it would be impossible to, sort of like, screenshot it or do anything as mundane as that because it's too big."*
>
> Participant 2: *"Oh sure. Sure. But every bit of digital data can be-, if it can be moved, it can be copied. Everything that's on the screen*

right now that I'm looking at could be copied, could be recreated, anything." Participants, Southern online group

Linked to concerns about data security and storage, participants also highlighted the importance of the vetting / due diligence procedures used to check individuals and organisations before they are granted access, including Genomics England staff, and how decisions are made regarding access (e.g., ethics committee).

"Yes, well, I think, for the parents, you need to give them the reassurances that, 'We've done the research into the people accessing the data,' you know? 'We've passed things through ethics committees,' you know? 'We've done due diligence before anybody gets anywhere near this data.'" Participant, Northern online group

A governance structure in place

Participants wanted to know that there is a governance framework managing all aspects of the programme, including discovery research. This needs to give clarity on what is expected of the researchers accessing data, what is and what isn't allowed, and if Genomics England has any redlines. For example, that data cannot be sold or shared, whether data is shared with international bodies and how this is controlled, and which organisations are not allowed access to the data e.g., insurance companies, the police and government.

It would be interesting if, like, they had firm boundaries, like, 'It will never be used for this, it will never be used for that.' Participant, London in-person group

Participants strongly argued that it needs to be clear what procedures are in place in the event that something goes wrong (e.g., a data breach), including being told what the worst-case scenario might be and how this is being guarded against. They also mentioned the importance of transparency when things go wrong and explaining what is being done to address the situation. For example Genomics England/ the NGRL should start by contacting the individuals who have been impacted by the incident before communicating about it more widely.

"Because most companies never admit a data breach unless it's made public, whereas the company that came to you that announced totally unsolicited that, 'We've had a data breach, and we are dealing with it, and here's what we've done to fix it,' is more acceptable than someone whose company got a data breach but try to hide it, and someone's actually picked it up." Participant , Liverpool in-person group

Participants also expressed concerns about how the programme operates in the context of areas of the NHS being privatised. Participants were concerned that in such a scenario, data could be used for solely profit-making purposes.

> *"If we're saying it's owned by NHS [...] it's realistically half run by government [...] and then other side can be privatised. So, who owns that information then? And who has access? Because that, sort of, terms and conditions of that situation suddenly change, because who you gave your data to isn't actually suddenly the owner of it."* Participant, Southern online group

This feeds into a broader concern felt by some participants from the beginning of the dialogue that life science and pharmaceutical industries are motivated more by profit than public good. They saw this as a potential risk in allowing researchers from these organisations access to the data without an additional layer of stringent approvals to check their motivations. Additional checks might include interviews with researchers to understand whether their sole motivation is financial, and to ask explicitly about the public benefit they expect to see as a result of their use of the data.

Some participants were concerned about discovery research because of concerns they held about the health ecosystem. For example, these participants felt that pharmaceutical companies profiting from people's illness is unacceptable in any scenario. They feared that discovery research into genetic conditions would be focused on developing treatments, rather than looking for cures, so as to make profits for pharmaceutical companies. Others thought that even if new treatments are discovered, participants might have limited access to them through the NHS due to cuts in funding. Some participants believed that society has become over-medicalised and think discovery research could exacerbate what they believe to be an existing problem.

> *"What, so there is now a medication for people born with the now identified CF gene? Perfect, you are now a lifelong customer of the pharmaceutical industry! If care for each individual was of the utmost importance, the pharmaceutical industry would stop using all this money in the cyclical motions of research, development and health and simply medicate for the cure."* Participant, Recollective

> *"Also with costs, I think people are going to be, "'I've got this particular disease now and I can't get drugs because you're telling me they cost too much. So why are you looking for more diseases that you're not going to be able to afford to get drugs for either?'"* Participant, Online Northern group

Participants in Liverpool felt that the idea of preventive medicine could be challenged if it seems to be motivated by profit. They discussed their sense that people who show a genetic disposition to a condition, but have no symptoms, might be unnecessarily put on medication. This they feel feeds in to a profit motivated system with no particularly useful research outcomes.

> *"If they're saying that they can be treated effectively before they show symptoms, it means that they, ideally, may want to put people on medication for something that may never happen or may never occur."* Participant, Liverpool in-person group

One group of participants expressed concern that discovery research might be impacted by the ability of participants to withdraw their consent at a particular age. They felt strongly that large numbers of young people are likely to want to remove their data from the NGRL when they are given the ability to do so, which would heavily impact the opportunity of discovery research.

Some participants referred to the possibility of unintended uses of newborns' genomic data which goes far beyond discovery research. These included biological warfare, genetic engineering, cloning, population control and being set up for crimes by criminal justice institutions. These concerns are often accompanied by references to science fiction and thriller films.

> *"Certain unscrupulous nations could use this information for biological warfare."* Participant, Recollective

> *"It might sound crazy, a bit unlikely, but ultimately, don't clone my baby."* Participant, Southern online group

3. Discovery research scope and impact

Participants expected that discovery research using newborns' data would focus on identifying rare diseases and finding ways for earlier diagnosis and treatments to be delivered. A number of participants also mentioned using genomic data for healthcare planning.

Participants were less commonly aware of the possibility of research for personalised medicine or increasing drug efficacy and making new drug development a more efficient process. However they tended to feel positively about these purposes when hearing from specialists and watching videos of researchers describing their work to these ends.

3.1 Impacts of research

Participants frequently referred to how the findings of discovery research undertaken using newborns' genome data might be used and the real-world implications of those uses. Participants shared a view that the positive impacts of genomic discovery research could include:

- research taking place which would otherwise have been difficult or expensive to do
- saving money on diagnostic pathways for future patients
- improving resource planning for demand in the NHS, or the education system, by understanding the likely prevalence of conditions in the future
- encouraging more funding for under-researched conditions
- enabling the UK to become a world leader in genomics, which they felt would be a boost to the UK economy
- helping to develop drugs which could benefit those in less economically developed countries

> *"NHS planning potential. Because if you know that there's a lot of people who are [predisposed] to strokes, Parkinson's or whatnot, then therefore you can be looking at long-term planning, can't you?"*
> Participant, Liverpool in-person group

However, some participants were concerned about the potential for other uses of research using newborn genomes' data. For many, these 'downstream' uses of the findings were equally important to the original purpose of the research, even if the researcher who wanted to access the data could not necessarily control how the findings were later used by others (because the findings would be published in an academic journal, for example). Participants therefore expressed some concerns about how they thought that research might be used, including:

- that greater knowledge about genetic susceptibility to disease could result in some groups being discriminated against or limiting their individual freedoms
- that greater understanding of links between genes and conditions could lead to prenatal selection or the eradication of neurodiversity (for example in fertility treatment)

- that treating and curing all illness will unnecessarily prolong life and increase population, putting additional pressure on the planet's resources
- that the NHS will be unable to afford to offer treatment for any new conditions discovered

> *"I can get my head around that there would be lots of checks and balances in place for individual research projects. When that research has been done and it's been published, how is that then used and for what purpose? [...] Can the research eventually be used to, kind of, almost force people down certain lifestyle paths and have it where you have these huge social engineering projects whereby how people live their lives is being dictated by the data that's available?"* Participant, Northern online group

> *"You could be doing all this and it could cost a small fortune, we haven't got the money to actually put it into practice or do anything with it, what's the point? And then, should we even be doing it in the first place?" (Participant, Liverpool)*

3.2 Mission creep

Participants commonly questioned how far discovery research using newborns' genomic data should go. Upon hearing about some scenarios, particularly where a condition might be added to the 200 conditions initially screened for, or where maternal data might be linked to newborn data, some participants considered that the purpose of the Newborn Genomes Programme could easily expand. For some of these participants, this cams with the fear that the programme might expand in ways they could not yet envisage, even when they remained broadly positive about the aims of the programme. We speak more about reactions to specific scenarios in Chapter 4.

> *"There's the original 200 [conditions being screened for] but yes, as we know, this is an area that will move on quite rapidly over the years. So, so long as the trust goes back in terms of how these diseases are being identified and how the data is then subsequently used to treat these diseases, but I think this goes back to the whole trust, in terms of making sure that there isn't just mission creep, that these things are being done in a thoughtful and structured*

way." Participant, Online Northern group

Participants also feared mission creep within individual research projects, feeling that researchers might obtain access to data in the NGRL for one purpose, but then use the data for another purpose (either maliciously, or as their research evolved from initial findings).

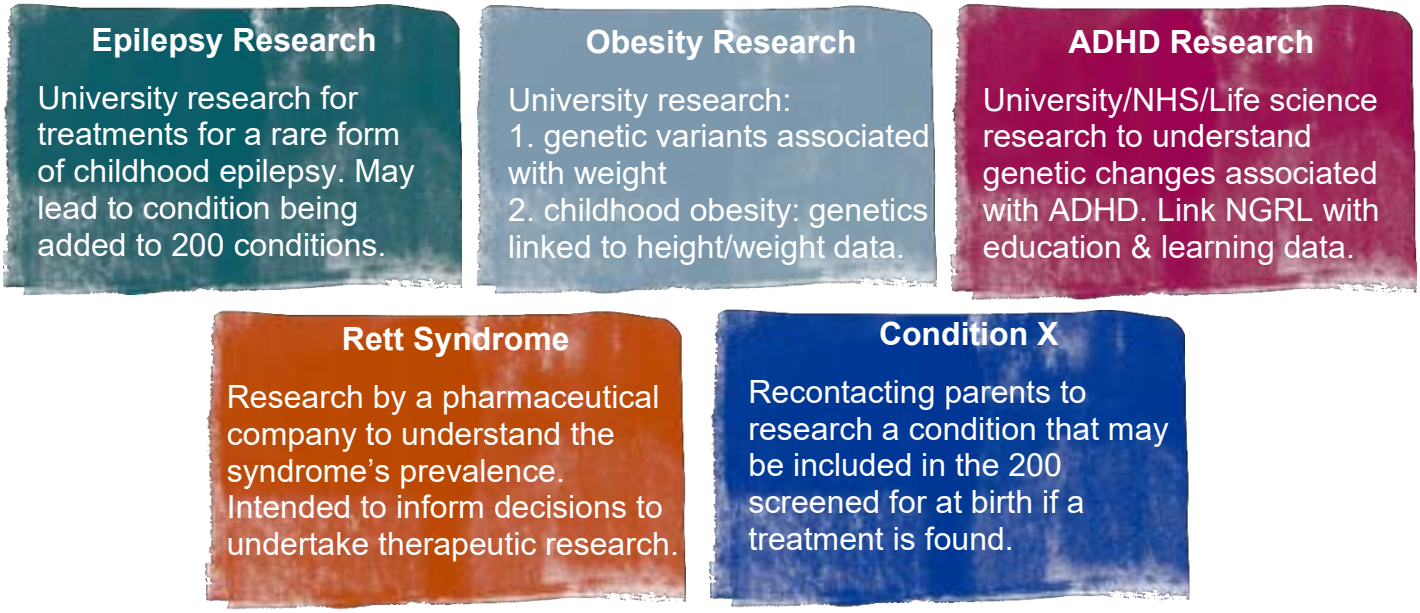
> *"So, if they're stating that this is what they're going to do, they have to ensure that, like, the process is covered and not kind of going in there and getting, I don't know, other information that they don't need. And then, basically, using the information that they stated they would use, for other purposes"* Participant, London online group

4. Research uses in relation to our scenarios

In this section, we draw together the key themes that emerged from participants' discussions about five scenarios. The scenarios were developed to stimulate discussions on different dimensions of potential discovery research. For example, whether the researchers worked in academia, the NHS and/or the life science industry; a range of types of conditions: physical and neurodevelopment conditions; childhood and adult onset conditions and a range of reasons for accessing the NGRL, such as to understand a condition's prevalence, to contact potential research participants to test a treatment, or to explore if particular genes are associated with a condition.

4.1 Responses to the scenarios (SK)

The five scenarios are:



The full scenarios are shared in Appendix A. The hopes, concerns and issues raised as a result of the discussion are set out in the following sub-sections.

Responses to data linkage within the scenarios

Linking external data to NGRL data was broadly welcomed by participants, provided it is proportionate to the research question and re-identification is avoided.

For the obesity scenario, the proposed external data to be linked was seen by some participants as insufficient. Obesity was seen as a complex issue related to many different physical, social and environmental factors as well as a potential genetic factor. The research described in the scenario is only asking for height and weight. Some participants think that far more data would be needed to do robust research into obesity that can achieve meaningful findings. This might include location and socio-economic data. However whilst they could see how necessary this range of data might be, it raised questions about how this data is incorporated, held securely

and not shared inappropriately. It also raised questions about the need for other family members' genetic data and some felt this might be a step too far in terms of data privacy.

> *"All they say is we're going to get a genome and then we're going to get height and weight and that's all the information you're taking. It's like me looking at your driving licence and telling you I know who you are as a person."* Participant, Southern online group

Some participants would like this kind of complex research mapped out in some way. They would like researchers to illustrate research scenarios that show which avenues involving which data they would pursue based on the incremental results they find.

Many participants believed that data linkage needs to be longitudinal, particularly for conditions such as obesity that potentially have a mix of environmental, social and genetic factors.

> *"If maybe 10 years down the line we don't link that future data with it (genomic data) to see what really happened with the kid, we don't really know how important that is. So linkage is how I think the research is going to go forward."* Participant, Liverpool in person group

Linking NGRL data to school and education data was seen as helpful by most participants for the following reasons - to:

- Learn over time what might be effective treatments for a condition such as ADHD
- Explore the efficacy of not just medical treatments but also social/educational interventions.

When the condition is not on the list

For the most part, participants saw the NGRL as a resource for improving health across the board, not just for the 200 conditions that would be screened for at the start of the NGP.

Indeed, for some, the most important benefit stemming from the discovery research element of the NGP is to better understand lesser known conditions, find treatments for them and add them to the list of conditions screened.

> *"This is really what I would hope to have got out of this database, that people can access it because they're looking at a cure for something. This scenario is everything I would actually hope from this whole programme, to be honest, that it's not just*

*the 200 things that we know about, but it's other things that they can find and help people."*Participant, Northern online group

Participants emphasised that, when parents are asked to consent to their baby's participation in the study, it is important to be clear that the discovery research element of the Newborn Genome Programme extends beyond the 200 conditions screened for at birth.

Participant Prioritisation

Participants had time and space to review and discuss the five scenarios and their relative merits across the dialogue workshops. This section summarises the factors they used to prioritise different types of discovery research.

The severity of the condition: conditions that are life limiting or with severe ongoing impacts, such as Rett syndrome or epilepsy, were a high priority for discovery research for participants because finding treatments or earlier diagnosis will have significant benefits for those with these conditions and their families.

Clarity of symptoms and potential for cure: a condition such as epilepsy was seen as having clear symptoms and if research offers the possibility of curing those clear symptoms before they take hold, this was seen as high priority research.

*"Of the three medical conditions presented I felt the epilepsy investigation would be of huge importance, especially to see if it's possible to cure before symptoms present. It also has clear symptoms and can't be disputed or questioned, like ADHD or obesity for example."*Participant, Liverpool in person group

In contrast, obesity was seen by many participants as inaccurately measured by the Body Mass Index (BMI). Given people's different heights, shapes and muscle mass they thought that trust in BMI was undermined when e.g., rugby players are diagnosed as obese. So, they ask, how can a genetic cause of obesity be determined, if we don't have an accurate way of measuring it?

"I think it, kind of like, boils down to the question of how is obesity measured? Because obviously I know in terms of BMI and things like that, but everybody has a different body shape."
Participant, London in person group

Potential to lead to an earlier and more definitive diagnosis: this was seen as a strong research benefit of the ADHD scenario. Several participants spoke of their own or family members' long journey before a diagnosis and the negative effect this had on their lives. Participants also indicated that a genetic link to ADHD would also have the important benefit of earlier diagnosis for women and girls who are often diagnosed late or not at all.

Clear potential for research leading to earlier health benefits, particularly in

childhood: for example, faster diagnosis and more effective treatment of epilepsy and ADHD leading to better outcomes for children e.g., fewer seizures. Such a route to earlier health benefits was less clear to participants for the obesity scenario.

Preventing misdiagnosis and unnecessary medication: the ADHD scenario prompted many participants to describe the benefit of avoiding misdiagnosis and therefore preventing the overmedication of children.

Potential to create personalised medicine: participants hoped that genetic research into conditions such as epilepsy and ADHD could lead to more personalised treatments with fewer side effects.

Informing what is known about conditions: participants valued research that had the potential to advance knowledge on a wider range of conditions than just the one being studied, for example, rare epilepsy research helping to inform our understanding of, and treatment for, dementia.

Helping to treat a condition in older children and adults: there was a strong hope amongst participants that research using newborn genome data could help both children and adults already living with a condition, such as epilepsy.

A truly genetic health condition: research using the NGRL has more importance and credibility in participants' minds if the condition is largely or solely genetic. When discussing obesity, some participants did not see it as "a genuine medical condition".

A medical condition or a socioeconomic/ environmental condition? Many participants felt conflicted with the obesity scenario. They recognised its prevalence, its impacts on people's health and its cost for the NHS. The genetic component of obesity seemed weaker than in the other scenarios, however some participants reflected on this and said this research could be valuable in proving this assumption wrong:

> *"I find this a strange one because I just suspect that the environmental factors and socio-economic factors are so much more a contributory factor than genetics, but maybe that's the purpose of the research, it's actually establishing whether that really is the case, maybe we've all been wrong, maybe actually it's all to do with genetics."* Participant, Northern online group

Some participants were concerned that defining obesity as a genetic condition could medicalise it and therefore reduce the imperative to address societal factors such as poverty and trauma.

> *"If you live somewhere where everybody you know drinks, heavily smokes, eats crap food, and the only place you can get stuff to eat in your local community is fast food outlets etc. that's what you're going to do... you could probably make a bigger difference*

in trying to address those issues rather than looking at genetics, I would presume." Participant, Northern online group

A condition where there are no or limited treatments or cures: participants identified conditions such as epilepsy and ADHD as having some existing treatments, but commented that many of these were not fully effective and/or had unpleasant side effects. In contrast, obesity was seen to have well known interventions in terms of diet, lifestyle and medical treatments.

"So there is a rich source of information out there with obesity, especially with healthy eating for children with school dinners, so there seems to be a lot of information already out there that makes you question as to whether we need genetic information on newborn children." Participant, Northern online group

Research that can inform both medical and social interventions: the ADHD scenario was for some participants a lightning rod example of a condition that could be over-reliant on pharmaceutical treatments. Their concerns were that the NGRL could be a bonanza for the life sciences industry and reduce the focus on social interventions such as specialist support as school.

"I think ADHD is kind of notorious for pharmaceutical companies using the condition for their benefit... I've known people who have been on a cocktail of drugs since they were very young, and it's easier for everyone else to deal with. And I think that was what was appealing about them mentioning getting support at school, because that implies that there's going to be some help with people who should be responsible, like teachers, being more involved in taking care of these children's needs." Participant, Southern online group

Reducing NHS and social care costs: research that could lead to significantly reduced costs for the NHS and social care services was highly valued by participants.

"The only positive I have in my head is obviously, obesity is impacting the NHS quite a lot financially. So, figuring out the genetic reasoning behind it would be good in terms of cost saving measures, but that's the only positive I can think of." Participant, Northern online group

How rare or common the condition is: the epilepsy scenario focused on a 'rare form of childhood epilepsy' which raised the question for some participants: shouldn't research be prioritised for conditions that affect a larger number of people? However, as discussed above, most participants saw severity of the condition and its potential to be treated early in life as more important factors for prioritisation for discovery research. The prevalence of obesity and ADHD in society was seen as the main benefit of using discovery research to understand their causes better.

Data access by life science companies

The scenarios that triggered discussions on life science companies involvement in discovery research within the NGRL were ADHD, Rett syndrome and obesity.

In the ADHD scenario, the researcher had a role in all three sectors: the NHS, academia and the life sciences industry. This was regarded positively by many, but not all, participants, because the researcher was seen to have a well-rounded view on the condition. They treat patients directly, have wider insights through their university work, and are aware of treatment innovations for this and other related conditions through their industry work. Where participants became uneasy in this scenario was when they thought the researcher could have an ulterior, financial motive for conducting the research. They wanted to make sure that the approvals process for those advising/ working in the life science industries is particularly robust. Participants want to ensure that researchers working in this context have a public benefit rather than financial motive for their work.

Obesity raised particular questions in some participants' minds. This extended beyond life science companies to include the food and diet industry. They wondered if the research could be used by these industries to target individuals with new products.

> *"What are they doing the research for? Is it for, like, a pill to help you lose weight? Is it a new, like, diet food?... I think it would have to be really careful that that data didn't go into the wrong hands, for commercial gain and stuff."* Participant, Northern online group

Research implications for the NHS

A few participants had questions about the impact that some discovery research could have on the NHS. They were concerned that some research could lead to recommendations for treatments before symptoms appear. They wondered if this could lead to over-medication for patients and excess costs for the NHS if, in fact, the genetic change did not always lead to a condition manifesting.

> *"I appreciate that with the DNA I suppose it is meant to guarantee that that is what it's going to do, but how do we know 100% that it will? Potentially we're giving people things that they might not even need and then the knock on effects of*

that, how is that medication going to affect them long term if they didn't even need it in the first place? And then also looking from the NHS perspective, you're putting a strain on the NHS which doesn't even need to be a strain because there's nothing actually there in the first place." Participant, Northern online group

Other points and questions about discovery research that participants thought should be made clear to those consenting to take part in the study are summarised here:

- How can newborn genome data help with conditions in adults? E.g. epilepsy
- Could a researcher just ask for data in the NGRL that has the genetic change they are interested in, or ask for everything?
- If a condition is rare, how can researchers be confident in your research if you can only test a treatment on a very small number of people?
- Can a genetic change be linked specifically to a condition's severity?
- Why do research on a condition using newborn data rather than people with the condition? Why not invest in more targeted screening/WGS of people with conditions with genetic links?
- What if someone doesn't have a genetic predisposition to develop a condition but they have all the symptoms? Are they going to be dismissed if genetic information is being used as part of that process?

4.2 A further exploration of responses to linking datasets

As well as reflections on linking datasets in the specific context of the scenarios, participants considered data linking in more general terms.

Participants views on linking data sets for discovery research can be characterised as 'broadly positive'. They felt this most strongly when told that:

- The approval process for research will be robust and rigorous and includes assessment by a research ethics committee (REC).
- Those consenting to take part in the Newborn Genomes Programme will be given enough information to understand that an 'all-in consent model' is in place, and they are consenting to data linkages for discovery research purposes as well as the screening element of the programme.
- The process of data linking is transparent and clear to parents and, as the newborns grow up, children and young adults.
- Accountability is embedded into the approval process with it being clear what the process is, and who is responsible if something goes wrong e.g. a data breach.
- Safeguards are in place to ensure that data linkages are always between sets of de-identified data and data cannot be re-identified using indirect or dishonest means, or inadvertently.
- Clear limits are set on the types of datasets that can be appropriately and ethically linked (for example, a red line for many is if linkages are made between Newborn Genome Programme data and crime or migration data)
- It is clear what the benefits of data linkages are expected to be for the specific

discovery research programme, and for wider society/ future generations.

An immediate consideration for participants was that linking datasets for health research presumably is not a new concept. They believed that learning from programmes which already do this should be applied to discovery research within the Newborn Genomes Programme.

> *"It's interesting to hear how different databases work together - it makes me think that perhaps there are already protocols in place to handle sensitive data such as this."* Participant, Liverpool in person group

Given the importance participants place on genomic and environmental data informing discovery research, many felt that linking datasets is an essential part of discovery researchers' toolkit.

> *"Sounds very important to have both genome and child health data to fully understand how these conditions work and whether they'll present themselves and how severely."* Participant, Northern online group

The benefits that data linkages could bring to discovery research were listed by participants as being:

- An improved understanding of genetic conditions
- An improved understanding of environmental risk factors for certain conditions
- A more fully rounded picture of the data from this largely healthy cohort of newborns
- A more specific understanding from genomic data by drilling down into richer data drawing samples from a range of sources
- Establishing longitudinal studies where greater insight could be gained from looking at linked datasets over time
- A greater understanding of the conditions that disproportionately affect some people in society e.g. sickle cell disorders and their impact on people with Black African and Caribbean heritage.

Overall having more data to work with and a larger sample than can be provided than by the Newborn Genomes Programme alone was seen as a good thing.

Whether or not datasets should be linked within the NGRL was seen to be a non-issue for some participants. These participants were comfortable with data linkages because the data is de-identified, because researchers have gone through an approvals process, and because they understand that specialist knowledge is needed to interpret and use the data.

> *"I actually don't care. They can have all my data because what are you going to do with it really?...Let's be honest, it's not a targeted thing, it's not about you. (Linking datasets) just gathers enough*

information to help others." Participant, London in person group

"For me personally, I've got no issue with it at all. If you're going through the vetting process in the first place, once that's done, if they are then linking it with other data then that's absolutely fine. I've got no issues. All the reasons for doing it seem pretty valid to me." Participant, Southern online group

For a few participants, linking datasets for discovery research brought some cause for concern. Two concerns that came to the fore both focused on deidentification.

The possibility of re-identification by linking datasets

Some concerns were raised that data can be more easily be re-identified if various datasets are combined. For example, if someone has a genetic condition, is being cared for within a particular Integrated Care Board (ICB) and lives in social housing, it might become more obvious who they are when reviewing the data. The more personal and specific the data is in participants' minds, the more uncomfortable the idea of linking data sets became for some.

"The minute you start bringing in height, weight, you're actually starting to pin down more specific details, rather than being, sort of, totally de-identified." Participant, Liverpool in-person group

The power of the data

For some participants, data has more 'power' when linked. Mostly this was seen as a powerful tool for public benefit with discovery research bringing new treatments, earlier diagnoses, and a clear public benefit. A few participants felt that this power could be used in a way which causes harm, including to manipulate findings for political or financial gain.

"It seems like when you are linking a lot of different datasets, someone who has access to all of them has a lot of power. So they could be doing a lot of manipulation in terms of what they can do with that data." Participant, Liverpool in-person group

These participants were also concerned about the spin that could be put on discovery research findings gained through linking datasets. They feared the media or industry could overstate or dramatise the findings.

"You can sensationalise the data almost and be like, 'Well we saw 20,000 sets of data with this gene, and when we combine that with data on weight and height it means that...' But that data from the

> *newborn doesn't necessarily relate to the data from children's height and weight. The data won't match up. It won't be telling you that."* Participant, London in-person group

It was important to all participants that data linkages are well-thought through and a clear rationale for needing the datasets to be linked for the purposes of their research needs to be articulated. This includes making sure that the right data is being linked to inform the research.

In this context, some participants felt that discrimination could occur by linking datasets, reinforcing prejudice in society. They wanted to avoid a situation where data linking allows bias or racial stereotyping to occur which would undermine trust in both discovery research and the main study.

> *"That one's truth. You are talking about trust. But they (the researcher) might have some bias or stereotypes which may affect how and why they put two sets of data together."* Participant, London in-person group

There was also a concern amongst a few participants that the process of linking datasets from outside the NGRL with the genomic data within the NGRL, could put the system at risk and cause a data breach. They felt that the more people involved in a process, the more likely the system is to break down. Reference was made to the Cambridge Analytica[2] scandal in this context.

4.3 Linking maternal health data

In the second part of the dialogue, participants were asked to consider how they would feel if maternal health data was linked to newborn genome data. Many consider this to be a useful approach, and a logical step, which could bring benefits in terms of data size, understanding the links between genetic and environmental factors, and having the potential for improved diagnostics, treatments, and therapies.

> *"I think that quite makes sense. I think it's a good idea, it's logical. It'd be good to be able to see if there were any links between perhaps the health of the mother during the pregnancy or any, you know, pre-existing conditions that the mother may have, how they then impact the babies genetically. So, this one, to me, sees like, logically, a good idea."*Participant, Northern online group

[2] From 2013 Facebook users personal data was collected without their consent by Cambridge Analytica, predominantly to be used for political advertising.

> *"I think it can have a positive correlation there, in regards to finding out certain information, patterns and linkages there. I feel like it can produce some useful information to research generally."* Participant, London in-person group

The belief that the consent process is important continues in this data linking scenario. Once in place, participants could see concrete benefits which could accrue from discovery research in this area. This included an understanding that health is not only about genomes and that environmental factors are also significant.

> *"Assuming that good quality consent has been obtained from the mother, this option gives a new layer to research. It lends itself to that nature versus nurture debate in a way. They'll be able to see understanding of, like, correlations connections things like that and it could help to understand is there any changes in the mother that cause changes in the genomes of the newborn. Yes, I think it gives a new additional set of data to expand the scope of what the research could do.* Participant, Southern online group

Benefits

Benefits to linking maternal health data with Newborn Genome Programme data were listed by participants as being:

- Links would provide a more rounded picture of newborns' genomic data and children's health
- Opportunities to link maternal health data with newborn outcomes
- The potential to give more insight into correlations between environmental and genetic factors
- Greater understanding of conditions which affect people from specific ethnicities
- An increase in the size of the sample, bringing more opportunities for discovery research at scale.

For most, this linkage did not change the nature of the Newborn Genomes Programme, but enhanced what could be achieved by discovery research using the data. Participants generally understood this to be a natural extension to the programme, bringing longer term benefits for future generations through discovery research.

For a few participants, linking these datasets might change the nature of the programme. They believed it added a layer of complication to the process which does not have clear enough benefits for those who take part.

> *"Essentially, they'll (mothers) just be guinea pigs if there's nothing*

to gain from it. So I think that raises more questions for me if the data's going to be collected but there's going to be no benefit to them in any way shape or form." Participant, Southern online group

Consent to linking maternal health and newborn genomic data

How consent is acquired for data linkage was important to some participants, many of whom wanted there to be a separate process for the mother from the newborn. They were also worried that an 'all-in' consent model might mean that the mother has to agree to share her complete health record, which she may not wish to do. They believed giving the option to limit the health record just to what has happened in pregnancy would be more acceptable to some mothers.

Reasons why a mother may not wish to consent included to protect her privacy. This was especially the case if the mother has events/ conditions in her history which she wishes to keep to herself and/ or she has reasons to be concerned about data misuse or re-identification.

"I heard of a case in hospital where a mother was expecting a baby, and she was HIV positive. Her partner did not know this, and it was not to be mentioned at any stage to the partner. So, there may be some people that don't want their information to be revealed."

Potential for harm in linking maternal health data

Who would see the data before it is de-identified and included in the NGRL for discovery research was also important for participants, particularly when there is sensitive material to be considered. Sexual assault or rape were examples given of information which might be known by a GP, but that mothers could feel uncomfortable sharing with others within the medical profession, or indeed Genomics England staff. This example links to the previous point where it might be acceptable for some, but not all, maternal health data to be linked.

"With data linkages, for example, someone who has been sexually assaulted or raped, or whatever, wouldn't feel comfortable disclosing their health records. Because you know, sometimes your GP records are linked to the hospital. So they may be comfortable disclosing it to their GP, but not necessarily to hospital staff. A mother wouldn't want to have that specific information disclosed, but she would like other information to be disclosed. But you know, sometimes, when you give consent, you give it blindly, and everything is consented, but

there is some information that they would like to remain confidential and separated from linkage. So that's the only challenge that I could see.

Equally, many felt that if mothers have already consented for their babies to be part of the programme, it is likely that they will want to consent for themselves.

"I'd also kind of question well if mum is happy for baby to do it, why is she not happy for her to do it? Like, that's just weird in my opinion if she said no for herself, but yes for her baby." Participant, Northern online group

Dialogue participants hoped that Genomics England would be in regular contact with the mother. They felt that a mother might change her mind about consenting to the linking of her own data during the course of the pregnancy and that those supporting the consent process should enable that to be a regularly stated option.

*"Constantly check with the mother that they were still happy throughout the whole pregnancy and after because I think hormones are like so high that I know if hormones are particularly in one way one day that may feel like, 'Oh yes, this is something I want to do.' But then another day they might think, 'Oh no actually thinking about it it's not something I want to do.'"*Participant, Liverpool in-person group

Due to their own or a family members' experience, some participants were concerned about causing anxiety or an adverse mental health reaction for the mother if, for example:

- Health records could lead to the mother being judged for where she lived, or how she lived, during pregnancy
- She feels under pressure to consent when she is fearful that the discovery research process might lead to re-identification
- She has a history of mental ill health which she believes leads to prejudice against her/ her child.

"When I was pregnant with them, I got put on suicide watch because I had really bad depression during pregnancy. I was grieving for my mum. Everyone's like, 'No, no you're going to hurt the baby.' So my head just went boof, so I had to go and see a therapist. Would that be prejudiced?" Participant, Liverpool in-person group

In summary, whilst many could see positives in linking maternal health data with newborn genome data, they advised that:

- Care should be taken to explain the process of deidentification and how researchers would use these linked data sets for discovery research
- Consent should be separate for mother and baby
- It should not be compulsory for a mother who wanted to consent to her baby being part of the study to have to consent herself
- It is more comfortable to consider linking pregnancy data, not the mother's complete health record.

"I don't think it should be linked to the whole person's medical records. That is too invasive. But that gestation period is part of the genetical make-up of the child, and because of environment as well. It's necessary for that period." Participant, Southern online group

Some participants had strong ambitions for the project and believe that this linkage should not be limited to maternal data. They believed there is a value to including paternal data in the study. They said fathers would feel disenfranchised from the process if their data wasn't collected too.

> *"You, kind of, feel a bit-, as I say, speaking in my personal capacity, as a dad, potentially disenfranchised from the whole thing."*
> Participant, Northern online group

They believed that increased sample sizes, data from fathers as well as mothers and linking environmental and genetic samples from parents could deliver greater insight from the discovery research.

5. Communications and transparency

Participants felt that open, honest, transparent, and accessible communications are essential to demonstrating the trustworthiness of the discovery research phase of the Newborn Genomes Programme. In discussing communications and transparency, participants did not separate out consent into the three aims. This is because of the 'all-in' consent model in place, where consenting to the screening element means that participants are also consenting for discovery research. We therefore start this section with the points participants made about communication and transparency in relation to consent, followed by communications with new and expectant parents, including on discovery research. We also summarise participants' views on:

- What expectant and new parents need to know about discovery research
- Communicating with wider society about discovery research.
- Whether and how to recontact families about a new condition.

5.1 Views on communications about consent

Consent is an important part of the study. Participants learnt that Genomics England would be using an 'all-in' consent model for the study. For many, hearing that participants could not just opt for the screening element, but would be asked to consent for discovery research too meant that the detail of consent should be an important focus for Genomics England's communication strategies.

What am I consenting to? How can I withdraw consent?

Participants emphasised the importance of giving informed consent to join the study, making it clear exactly what parents are signing up e.g., what their rights are; or what the risks of taking part in all elements of the study might be.

> "Yes, so, like, some parents, they should be given and told what the data is going to be used for. They should know, like, how it's going to be used so that they have a full understanding of what it could be used for and what are the possible outcomes of it. So, before they consent, they should understand what they're really agreeing to."
> Participant, London in-person group

They also emphasised the importance of information about all elements of the programme being easy-to-understand and accessible to all, particularly when using an 'all-in' consent model.

> "It needs to be done in layman's terms because the amount of people that just skip terms and conditions. Or, you know, they'll argue, 'I didn't know what it was being used for,' and they'll come back with, 'Well, you should've actually read.' Do you know what I mean? I

think people don't want to throw themselves into so much commitment when they've just got a baby on the way. It just needs to be really clear-cut instructions, and no hidden clauses, I suppose." Participant, Liverpool in-person group

Participants also indicated that it needs to be made clear how to opt in, but that families must feel under no pressure to take part.

"Well yes, yes as a parent, as a Black woman, when I was pregnant they automatically came to me and said, 'We want to test you for sickle cell anaemia, we want to test your partner, and then when your baby is born we're going to test your baby.' And I said, 'Hold on a minute, are you asking me, or are you telling me?' And that's when they kind of stopped in their tracks, I said, 'You can't automatically do anything without my consent.'" Participant, London in-person group

Participants also highlighted that parents, and the children as they grow up, must know they can withdraw consent at any point, and how they can go about this. It must be clear that they are "in control of their own data."

"I guess just knowing that you can give and take away your consent whenever you want at any point without any conditions that are difficult to jump through those hoops, especially for new parents who might not be able to navigate the process I suppose."

A few participants felt it might be useful if families could be included in the study on a rolling programme of consent. In this model, parents could consent to their newborns' data being used in some discovery research studies and not others. This, they felt, would provide further opportunities for clear communication about the programme in general, and discovery research in particular, over time.

5.2 How to communicate with new and expectant parents

Giving consideration to communications about the Newborn Genomes Programme in general is important for participants, and an integral part of the discovery research element of the programme. Participants agreed that good communications are an essential precursor for a successful discovery research programme. As such we include here points made about communications on the programme as a whole.

How to reach expectant parents

Participants recommended a range of targeted communications for expectant parents to encourage them to be involved in the programme. Having data for

discovery research is contingent upon people agreeing to be involved. They recommended a wide range of communication methods, from media advertising to direct contact via health professionals. One participant described it as building a brand that people trust, and to consider a family's journey from how they first find out about the programme. Another commented on the need to communicate in a way that builds awareness without generating fear.

> *"I think it must be, sort of, given to them in a way that elicits interest without causing undue fear in parents. Yes, because once the information is out there, then people will make their own mind up. It's got to go out to the public in a way that elicits, again, that sensation of trust somehow, you know, to come forward, to volunteer if you like."*

Mainstream media and social media channels were recommended to raise awareness of the Newborn Genomes programme, including the discovery research element. Sources mentioned included television (e.g., a documentary, news clips, relevant programmes, adverts), radio, podcasts, advertising on transport, everyday medical settings (e.g., GPs surgeries, waiting rooms), and social media, such as Instagram, Facebook, YouTube, targeted pop ups. Participant suggested that those interested in finding out more should be signposted to Genomics England's **website**.

Many participants thought expectant parents should also be approached directly by **healthcare professionals** who are trusted and have knowledge of the Newborn Genomes Programme, when they are at an appropriate stage in their pregnancy. This would be an opportunity to ask questions and learn more about Newborn Genomes Programme and discovery research. Suggestions included midwives, GPs, hospital doctors and other healthcare professionals. A few thought that there may be a role for Genomics England to answer questions.

> *"I think the one thing that keeps coming up is whoever is briefing anyone about this on the form, and anything about it, they have to have extensive knowledge of the programme and all related questions this person could have. Like, I don't want to hear about the programme while I'm trying to have a baby, basically. Then they go, 'I'll send you a link so you can read that.'"*

Many participants emphasised the importance of providing expectant parents **with tailored information**, which is human-centred, so that they do not feel overwhelmed when learning about the study. They talked of the need for face-to-face interaction, and being able to easily contact someone with knowledge of the programme if parents have follow-up questions.

Many participants saw value in there being **an information booklet / welcome pack** for parents thinking about joining the programme, provided by the health

professional.

> *"Perhaps when, you know, when people have new babies they often have, like, a welcome pack, don't they from the hospitals. So maybe there should be a leaflet that gives the initial information with a number or perhaps somebody from the programme could then visit the maternity units or maternity clinics, to say, 'When you have you baby, do you want to become part of this programme?' Or even have somebody from the programme have their own little office, you know, at the maternity clinic so that people can go and talk to them if they want to."*

Participants also thought information should be provided in other formats, such as **video or audio**.

> *"I was thinking in a similar way [video], will the process sort of be documented so people can see examples of the data being taken and sequencing and all those kind of things so they can actually see every step of the way what's actually going to happen? Maybe like little clips from parents and things like that who have opted into the programme and things like that so people can, I guess, relate to what their experience might be like if they choose to opt into the programme."*

A few suggested **sending a letter and information pack directly** to expectant parents who may be eligible to take part directly, for example by using a filtered NHS dataset.

Some participants discussed when an appropriate time is during pregnancy to talk directly with parents about the possibility of signing up to the NGP / discovery research.

> *"Discussions at different stages. So initially, very early, and also when they're closer to end of term because that's the time that your, kind of, absorbing more information."* Participant, Southern online group

5.3 What expectant and new parents need to know about discovery research

We now focus on communications about discovery research. Participants felt it is important for families to know who has requested access to data from the Newborn Genomes Programme, and who has been granted access (e.g., universities, pharmaceutical companies). They wanted to see a list of all research studies, with full transparency on each, including the project purpose, how the data will be used and what data linkages will be made. One participant suggests having a timeline, such as they have seen for the Human Genome Project[3].

Participants argued that having up-to-date details about discovery research studies in the pipeline will support informed consent for future participants and provide further opportunities to remind families that it is possible to withdraw consent.

> *"It's that transparency you would want to know as a parent, what research that data is being used for. Partly out of interest, but also if you then felt uncomfortable with anything, or you were questioning how it was being used, then you might want to withdraw consent. It's keeping people informed, you don't just give consent and then forget about it for 18 years, you need to feel like you're actively being kept updated."* Participant, Northern online group

Knowing what discovery research is being conducted also gives further opportunities to sharing positive news stories about discovery research. Participants highlighted consistently the importance of doing this as part of a package of activities to:

- Promote the programme and its work
- Explain what discovery research is
- Demonstrate the positive outcomes that come from discovery research, even if those benefits will accrue for people in time to come rather than immediately.

> *"If you donate blood, then a couple of weeks later they'll send you an email saying, well the one I had most recently, 'your blood has gone to Leeds General Infirmary,' for whatever purpose, so if there was something like, I don't know, like a monthly newsletter, or a quarterly report of good news stories, good outcomes that had come*

[3] An international project which worked to sequence the human genome: https://www.genome.gov/human-genome-project/timeline

from the data. So, 'Your inclusion in this research programme has enabled whoever to develop whatever,'. We're just keeping people in the loop as much as they want to be." Participant, Northern online group

Some commented on the need to share both positive and negative research outcomes.

What does taking part in discovery research involve?

Participants raised a number of other questions expectant parents are likely to have when considering being involved in a study that includes data access for discovery research:

- What are the benefits for me and my baby? Do I benefit from the discovery research?
    - Participants felt it is important to make it clear that direct benefits might not accrue to those who sign up to the programme but might be more for future generations as the outcomes of discovery research are realised.
- What are the risks and dangers of taking part? Could the data and/ or the discovery research findings be used in a bad way against my child in future?
- What level of commitment is expected once you are a participant, what will happen in years to come?
- How often will I be contacted? What might I be contacted about? What is the procedure for recontacting participants?
- What support is available if participants need advice at any stage (e.g., counselling, psychologist) whether or not their baby has a positive result or not?

How to communicate about discovery research once families are i participants in the Newborn Genomes Programme's study?

Knowing about the study was felt to be essential. Knowing about the discovery research element was highlighted as equally important. Participants emphasised the need for proactive, ongoing communications with families who have consented to take part in the study, so that those who are interested in keeping abreast of discovery research do not feel "in the dark." Some participants wanted to see a dialogue between participating families, Genomics England, and discovery researchers.

Participants recommended a few keys ways to communicate with those taking part:

- An **online portal, an app and/or a website** that keeps parents abreast with what discovery research is being conducted, that they can access as and when they want to.

I think it might be nice for parents to be able to, kind of, see a bit more about what's going on. So maybe have, like, a parent portal

that you could log into that showed the type of access requests that were being made, just, like, as a brief summary. So for example, 'A pharmaceutical company request access to X, Y and Z for the purpose of X, Y and Z. Information linked in with this would be height, weight, etc., but will be de-identified as always.' So just, like, maybe a brief summary. Participant, Northern online group

- A **newsletter** that provides periodic updates on the Newborn Genomes Programme and discovery research.
- A **forum (online and/or in person) for parents** where they can ask questions about discovery research being conducted and have a dialogue with Genomics England. Participants felt that this would be useful for families who are considering taking part as well for those who are active participants.

"I do think these kind of town hall meetings that I mentioned the other day, they could be an opportunity for those, as well. They can be done (online), just an opportunity for people to be able to maybe have that more personal opportunity to hear what's going on and ask questions etc. that kind of thing." Participant, Southern online group

A few participants worried how a family whose baby has received a 'condition suspected' result for the screening element of the programme might be impacted in the event that discovery research sheds new light on the suspected condition.

"Some of the information that they were talking about, information that can come out (because) of medical papers that are written, though it might have anonymised details on it, it might provide further information about a newborn's condition that might be upsetting. If the parent just finds that their child has a particular condition, and then research papers are written, and if the outlook is bleak, that could be quite negative for the parents to find out." Participant, Recollective

Communicating with wider society about discovery research

Participants saw value in wider society being aware of the NGP and discovery research being conducted. Proactive communications and education will help to raise awareness, demonstrate trustworthiness, prevent misinformation, and "put minds at rest."

> *"I put 'provide education and information on the research and outcomes to dispel any myths and misinformation'. So, the more that they get their information out there, the less likely it is that other people will be able to manipulate it for their own use, you'd hope."* Participant, Northern online group

Some participants argued that all information available to expectant parents should be accessible to wider society. Others commented that those not directly involved in the programme will need less detail and reflected that some people will not be interested at all.

Some felt that not all members of the public will be supportive of discovery research being conducted. Some argued that there needs to be careful messaging to ensure that the Newborn Genomes Programme is well received; others commented that there will always be some who are critical and emphasise the importance of transparency and proactive communications.

> *"I think it needs to be recognised. There's always going to be people who will positively see that and there'll be people who will negatively see this in different lights. And society is made up of a whole range of, you know, different types. So we need to appreciate, or Genomics England needs to appreciate, there is always going to be a negative view in the community as well as more than likely a larger positive view. How do you win over those negative views?"*Participant, London in-person group

Some participants felt that macro societal, ethical, and medical considerations needed to be discussed and debated with wider society, just as was explored by participants in this dialogue. For example, whether certain types of discovery research raise ethical considerations, such as obesity research.

> *"I feel like wider society may have more interest in the wider social political medical implications and just as a whole, because they maybe won't be as concerned about the individual risks and individual benefits because they're not involved in the project, but they may want to know more about how is this actually going to impact wider society on a more macro level?"*Participant, Southern online group

Participants commented that Genomics England needs to be better known and understood by wider society, if people are to consent for its study and for discovery

research to be accepted. Some recommended promoting Genomics England as a whole, rather than focusing solely on the Newborn Genomes Programme's study and discovery research. Participants thought that the involvement of the NHS needed to be prominent in communications, given it is a known and trusted brand.

> *"I think you'd need to gain people's trust by, like, literally putting NHS on the advertisements because otherwise people might think-, you jump to conclusions like, they want money out of you or they're not trustworthy."* Participant, Liverpool in-person group

Many of the suggestions about how to communicate with wider society about discovery research were similar to those suggested for expectant parents. For example, a media campaign using various channels e.g., TV, newspapers, social media channels such as YouTube, Facebook, and a website.

> *"We were talking about the constant updating of the website. If anything new was discovered or established, that data would be updated on the website in layman's terms, so the wider society can understand what the research was, why it was conducted, and all the research of the researchers."* Participant, Northern online group

Other suggestions included featuring the study on popular science programmes. One group discussed their disappointment that 'Tomorrow's World' is no longer on TV as they felt it would be a perfect forum to share this. Others discussed documentaries and televised debates that address wider ethical and social considerations raised by discovery research.

Participants emphasised the importance of trusted individuals, who are good communicators, delivering messages about the research, such as medical professionals. Specific mention was made of Professor Chris Whitty, Chief Medical Officer (CMO) for England, the UK Government's Chief Medical Adviser.

Participants recommended that people who have taken part in the study should be involved in communications. One person suggested a documentary that follows some of the children participating in the study over several years, like documentaries 7 Up[4] and Child of Our Time[5].

[4] First aired on television in 1964, produced by Granada Television, the 7 Up collection followed the lives of 20 children from age 7 to the age of 63.

[5] Produced by the BBC with Professor Robert Winston, Child of our Time follows the lives of 25 children born in 2000.

5.4 Whether and how to recontact families about a new condition

During the dialogue, participants discussed a scenario which explored recontacting participants about a clinical trial to test a potential treatment for a genetic condition that was not part of the original 200 conditions. In this scenario, families of children with the condition would be contacted to make them aware of the condition and to invite them to take part in a clinical trial. The scenario generated discussion and deliberation amongst participants about whether and how to contact families.

Should participants be contacted if a new condition is identified?

Some participants thought that there is a duty of care to inform families if discovery research sheds light on a genetic condition that their child has, and which they were not previously aware of.

> *"I think it's a good thing, because the reason you signed up to this research in the first place is to help the child get diagnosed earlier. So if new things are coming along, I'm sure they want to be diagnosed earlier with them, as well."* Participant, Southern online group

Some participants thought it important to let parents know about new conditions that could be revealed through discovery research. However, they felt this is contingent on it having been made clear to parents at the time of consent that the programme may add new conditions that are treatable. Others felt there should be a choice at the consent stage whether parents sign up to being contacted about new conditions which go beyond the original 200 conditions. Some also suggested that there should be a choice to be contacted a) if there is research into a potential treatment or b) only when there is a confirmed treatment.

> *"Maybe there should be something in that initial policy document or whatever that says yes, I want to be re-contacted that says I only want to be contacted if there is a clear treatment available or a care plan available. And then a separate consent to I want to be contacted if it's for further research to develop."* Participant, Northern online group

> *"Like, if there's certain conditions that a parent would rather not know about and obviously, if the whole genome's been analysed, the researchers will find that and it's just, like, making sure that stays confidential and whatever the parents would rather not know, they don't know."* Participant, Liverpool in-person group

Some stressed that parents should not be informed if there is currently no treatment or care pathway for the children affected.

One person reflected that it may not be possible to inform the parents under this scenario because it is a clinical trial, and that there would need to be a mechanism to quickly add new conditions that are identified through discovery research to the 200 conditions.

> *"... how quickly could it [the new condition] be added and can that then be used retrospectively to flag up to those affected of the families of those affected newborns and I don't know if we know that in terms of how that might happen. I suppose that's what we're doing with this really isn't it but, yes, I would agree. You would like to think that there might be that mechanism where they might very quickly be able to add it to the list and for it to help those individual families."*

How should participants be contacted if a new condition is identified?

Participants had differing views on what would be the best way to recontact families under this scenario. Some thought it should be Genomics England because it is responsible for consent and should have a team who are trained to deal with these kinds of situations.

Some commented that it should not be the researcher in the first instance, because they are working with deidentified data and there could be concerns about data breaches.

> *"Because once you've allowed a third party to do that you've got less control over how they go about doing that and there might be concerns from the individuals being contacted. You know that initial reaction is woah, why am I being contacted by a researcher rather than Genomics England or the NHS are the people I have dealt with previously. It could start a lot of concerns running in my head about what's going on with data and all of that particular issue."*

There was also concern that researchers would not be skilled to deliver this kind of information because they may not be healthcare professionals.

Many thought that contact with families best rests with an NHS clinician, a genetic counsellor, or a specialist unit with the experience and knowledge to handle the disclosure well and support the family.

Views varied on whether it should be a person's GP. Some people thought GPs are

under too much pressure and would lack specialist knowledge. Others liked the idea of first contact being with their GP, given that they are likely to already have a trusted relationship with the family. It is therefore important to these participants that GPs are fully aware who in their care is part of the study.

Participants shared a range of views on what is the appropriate mode of communication in this situation. Many of them thought face-to-face contact would be important. Others saw value in information being available to re-read online because meetings can be overwhelming. Some worried that letters may not be received, others preferred the idea of a telephone call followed by a consultation with a specialist. Some also highlighted the need for communications to be accessible, for example having someone who speaks the participant's language. Discussions also focused on the importance of providing those who are contacted with support, someone who can listen to their anxieties and can tell them about the condition.

6. Considerations on trust

In this section, we set out what participants considered to be the principles of trust. We relate this to explanations of fairness during the dialogue. Our findings in this chapter end with:

- What participants expect of Genomics England in order to the trusted guardians of newborns' genomic data
- What participants expect of researchers, including those from the life sciences industries, when accessing data within the Newborn Genomes Programme.

6.1 Principles of trust

During the dialogue, participants were asked to describe experiences, people and/ or organisations that had demonstrated they could be trusted. In this context, people described trusted:

- **Relationships:** a friend or a relative, mothers, sisters, friends
- **People in the media:** such as. Martin Lewis or David Attenborough
- **Organisations:** such as the NHS, including specific NHS hospitals or services, third sector, networking and support organisations e.g. MIND, Age UK or Marie Curie
- **Private sector organisations:** some mentioned banks, other financial services, or a brand they trust.

> *"To trust means to rely on another person because you feel safe with them and have confidence that they will not hurt or violate you. Trust is the foundation of relationships because it allows you to be vulnerable and open up to the person without having to defensively protect yourself."* Participant, Recollective

Some participants spoke about situations where trust had been lost, for example a broadcaster they think normally does a good job in presenting the facts, but who then endorses a product which clearly is not good for society e.g. junk food or gambling, leading to mistrust in the areas where previously they thought them to be trustworthy. Many participants discussed the fact that it takes a long time to earn people's trust, but very little time to destroy it.

For a few participants, this exercise was challenging. They said that they do not trust anyone or any organisation. This was seen in the context of having been 'let down' or 'disappointed' by people, organisations and companies in their lives. One participant used the experience of Covid-19 to explain the mistrust that they felt:

> *"To be honest, during and after Covid, I don't trust any organisations , after the many lies the public were fed in order to*

push the vaccines, fooling the masses into believing they were doing it for the greater good, to protect others and themselves, and therefore joining human clinical trials under the guise of protecting their health. The world is an evil place. I am very sceptical these days and find it hard to believe anything is being done ethically."
Participant, Recollective

This led the groups into discussions about what trust means and revealed a series of 'principles of trust' that people apply when considering how trustworthy an individual, an organisation, or a system is.

The principles participants apply to a trusted person, organisation or service are:

Act with **transparency**, using clear communications and with the expectation of openness in all relationships.

Define, and act within, high **ethical** and **moral standards**.

Be **reliable** and **proactive**: say what you are going to do, do it, and tell people that you have done it.

Be **genuine**: show that you care, you are empathetic, and you will stand up for what you believe in.

Be **discreet**, what I share with you shouldn't be shared with anyone else unless I've agreed to it.

Put **safety** first. Do nothing to harm people or knowingly put them at risk and have safeguards in place for when things do go wrong.

Demonstrate that **public benefit** comes before financial motifications.

Commit to a **mutually beneficial relationship** and be clear about what both sides are committing to.

Of these, 'transparency' was the principle that came to the fore as highly significant for trusted relationships.

> *"When my mum was diagnosed with breast cancer, and with the meeting with the consultants, they were very truthful, and they told us that there wasn't any more treatment that they could do. They told us it was terminal and she'd have months to live, not years. So they were clear with us with everything. I think that's that kind of transparency, from the start, I think."* Participant, Northern online group

6.2 What is expected of researchers

The principles applied to researchers highlight participants' desire to know that those gaining access to newborn genome data have been through a rigorous approvals process. They wanted to know that those who are working on the data are working within strongly defined ethical frameworks and have public interest motivations.

> *"I am all for researchers such as Pirmohamed*[6] *having access to gene databases to create better pharmaceutical solutions. However, protocols, protections and ethical committees still have to be in place if you're creating a database of this kind."* Participant, Recollective

This need for concrete assurances on the approvals process being in place and being robustly applied was rooted in fears about researchers' past actions. Ensuring only those who work within the highest research standards can access the data is essential to the trustworthiness of researchers and the research process.

> *Participant 1: Again, it's just, kind of, keeping the research to a confined group of people because anybody could say that they're a researcher and they need to carry out this research, and their intentions might not be genuine and pure kind of thing.*
>
> *Participant 2: There's many instances of researchers going and vaccinating in certain places around the world because they are considered third-world people, and they've died because they're killed*

[6] Professor Sir Munir Pirmohamed, Professor of Pharmacology and Therapeutics, University of Liverpool spoke to the in-person group in Liverpool, the recorded presentation was shown to all participants on Recollective.

them, basically. Participants, London in-person group

The factors participants wished to see embedded in this approvals process included assurances that researchers:

- Have the right skills, experience and training – i.e., they know what they are doing when they are undertaking discovery research using the data.
- Can work with probity and having nothing in their past, including a criminal record, which might put this into question.
- Understand that their approval will only give them access to the data necessary for their research and no more.

"(Working in the civil service), when we were accessing the client database, I could, say, if I wanted to, look up and find out about my neighbours' problems with the kids or who's got a disability, all this. So firstly, I had to have a password for it, secondly you're only allowed access to particular datasets. So it's access rules and passwords and all those practicalities. You know, when you sign onto something, it should say straight away,'" Participant, Liverpool in-person group

- Can demonstrate that they have a robust research plan and have a clear rationale for the need to use newborn genome data.
- Have declared any conflicts of interest and those managing the approvals process are satisfied that any such conflicts have been risk assessed.
- Have shown how they are funding the research, and this is tested to ensure that the funding source is appropriate and in line with public benefit motives.
- There is a peer review process in place for the discovery research.
- Are committed to sharing the findings in ways that can be understood families who have consented to be part of the study, and wider society.

"I think Genomics England should say, 'Please share your results,' that's what I was saying before. Share your results with us, you know? This is a stipulation, once you find out, let's take this further, let's share the information." Participant, Liverpool in-person group

Participants throughout the dialogue referred to newborn genome data being 'sensitive'. We find no evidence that the sensitivity they refer to is related the data coming from newborns. Rather, that the data is genomic and is about a person's make-up, questions of identity, and 'who I am' genetically. For this reason, it is perhaps the most sensitive data of all, and should be treated with enormous respect by all those involved in the programme. This is also why the deidentification of data is essential to participants and researchers should not, at any point, be able to link data back to a specific individual as they conduct their discovery research.

"You have to do more checks than maybe you think are really necessary, I think, when dealing with something this sensitive."
Participant, Southern online group

Industry researchers

There is a sense that, in terms of trust, researchers from pharmaceutical companies and the life sciences industries are in a specific 'for profit' category. Participants felt that this should be recognised and therefore additional measures put in place for research which could be motivated more by return on investment and profit, than for public benefit.

Many said that it should be ensured that for-profit companies pay back into the Newborn Genomes Programme. By contributing financially to the programme, industry researchers could demonstrate that they value the data and recognise its importance.

*"I think it's difficult to separate pharmaceutical companies from medical research, as a lot of funding for this research comes from them. I think the important thing is that they have to financially contribute to use databases that are publicly funded."*Participant, Recollective

"A lot of researchers work for big pharmaceutical companies, and they make many millions in profits anyway. So if they want to access any information, they should pay for that information."
Participant, Northern online group

Assurances for these researchers should include confirmation that they are not selling the data on to third parties; and that there will be spot checks on the researchers to ensure they are complying with this, not only as they access the NGRL, but during the lifetime of their research. Principally, participants were worried about motivations. They wanted to see checks in place to ensure that financial considerations are not the sole motivation for wishing to do discovery research in this area. This interchange between participants is typical of the discussions in all dialogue workshop groups.

Facilitator: *So, what is it about that specific thing? What is the problem with him being an advisor to a pharmaceutical company?*

Participant 1: *Pharmaceutical companies stand to make a lot of money when manufacturing an ADHD medication. Which, I guess, could it be influencing this work?*

Participant 2: *They can have, like, a bias towards making an expensive medicine for ADHD.* Participants, London in-person group

Many participants felt that trust in pharmaceutical companies has been eroded by repeated practices which do not appear to have public interests at their core.

> *"Yes, also these corporations have a history of this behaviour. So that's what makes it hard to trust, I think. Like, a history of not being trustworthy, so in various ways I always feel very sceptical."* Participant, Southern online group

Participants indicated that paying into the system and having more rigorous checks on researcher motivations would help to demonstrate trustworthiness.

6.3 Demonstrating fairness will demonstrate trustworthiness

A number of participants expressed concern about institutional bias in healthcare settings and a lack of diversity in healthcare research. Participants saw this as fundamentally unfair and a reason for loss of trust. They emphasised the importance of ensuring that this study is different, and that both the programme and the discovery research undertaken within it, reflect the diversity of the population.

> *"I think I'll say that there are obviously certain disparities when it comes to healthcare research with different minority groups. And whilst healthcare research is good, as a whole, in terms of the participant and the research done, it doesn't always reflect the whole population of the UK."* Participant, Southern online group

Some highlighted the importance of understanding how people from diverse ethnic and religious backgrounds may question the ethics of discovery research in the programme. They reflected on the impact people's experiences of suffering discrimination will have on their willingness to take part and the concerns they might have about using the data for discovery research.

> *"In regard to this pool of data that's going to come from a diverse background, with so many made up of different ethnicities and religious backgrounds. And those religious backgrounds, they'll have their traditional beliefs. They'll have their faiths. Some might feel that they might question the morals behind this, the ethics behind this. They already might have a feeling of being targeted before in terms of some form of discrimination in their life, you know, me*

> *being of a South Asian background, Muslim faith, I've suffered discrimination in my time, and I'm born in this country. Those are things that are ingrained so that could be a potential stumbling block."* Participant, Northern online group

Some worried that communities who already feel discriminated against may be further disenfranchised if they feel that Genomics England are "cherry picking" who takes part and whose data is used for discovery research. For example, one person understood that a cohort of 'healthy babies' would mean communities who experience high incidence of some conditions such as sickle cell disease could not be included in the study. This example highlights the importance of clear communications to avoid misunderstandings.

> *"Oh no it's just because it was actually in the text and that's what kind of rattled us a bit, they said that it's only healthy babies. You've got certain parts of communities in London, across the UK that they feel discriminated against, they think there's an imbalance in society, they feel that they don't get the same treatment as somebody else from a different background. So something like this could potentially fire the fuel for their thought process, with them feeling more the way they do. So that is why, but if you're saying that's not the case and if it's genuine."* Participant, London in-person group

Participants felt that fairness within the programme would be demonstrated if Genomics England is seen to be working hard to take an inclusive approach to:

- Who is included in the study
- Where they live
- The research that is undertaken using newborn genome data.

This relates to all minoritised groups within society, women, and people who live in disadvantaged communities.

A few participants mentioned their perception that current health research does not do enough research on treatments, medications and therapies which would improve women's lives.

> *"I said the thing about how there's often a lack of research in conditions that specifically target women, or how often medical stuff isn't researched enough into how it affects women differently.*

I think a perfect example is the pill, it has loads of side effects, and we don't always necessarily care about how that might affect women, we just kind of expect them to get on with it..." Participant, Southern online group

6.4 What Genomics England needs to do to demonstrate it is trustworthy

Given participants' focus on good communications and the high priority they place on transparency, it is clear that there a number of steps Genomics England can take to be recognised as a trusted guardian for this sensitive and valued data. These steps are rooted in the principles set out in section 6.1 and are set out below. Unsurprisingly, they begin with transparency.

1. Transparency is key to trustworthiness

Participants stress that transparency is a prerequisite of trustworthiness. They wanted Genomics England to communicate widely and consistently about the whole programme and discovery research. The ways of doing this that participants explored are set out in Chapter 5, but include:

- Regular updates on what discovery research is being done with the data
- Reporting back on research findings
- Explaining when things go wrong, or do not turn out as expected, as well as the success stories.

2. Raise awareness

Many participants said they were unaware of Genomics England and previous projects such as the 100,000 Genomes Project before taking part in the dialogue. They felt it will be hard to demonstrate that Genomics England is a trusted organisation, without first raising awareness of the organisation and its work first.

"Personally I'd not heard of Genomics England before I did this so I think if it was me, I'd kind of be going, 'Oh who are they? What are they doing with that data?' Whereas obviously the NHS is a big machine and it's well known and yes, maybe to educate people on who Genomics England are and what they do and all of the great stuff that they've done before this that we all now know about." Participant, Northern online group

3. Promote Genomics England's relationship with the NHS

Some participants felt that Genomics England would demonstrate it is trustworthy if its relationship with NHS is spelled out. They felt, particularly since the pandemic, that the NHS is highly trusted across society and if it were well known that NHS clinicians and other staff are involved in the programme it would reassure people that

Genomics England can be trusted.

> *"I think it's important to stress that this is in partnership with or in conjunction with the NHS because I think a lot of people might have negative views, but I think the majority of people are quite pro-NHS and I think that's a good sort of point to put across."*
> Participant, London in-person group

A few participants went further, and said that an efficient way of demonstrating trustworthiness would be for Genomics England to move under an NHS umbrella.

> *"We'll go to the doctor's and we'll let the doctor or the NHS store all of our information, very personal information. We don't think twice about it. But, actually, how does Genomics England get that same, kind of, trusted status with their data as with the NHS? I know I keep going on about branding, but, I think, you know, if this is such an integral part of the NHS in the future, why isn't it branded from the NHS? So they automatically own that and get that trusted status?"* Participant, Southern online group

4. Show your credentials

Just as participants wanted to ensure researchers have the right skills, experience and credentials to conduct discovery research with the data, it was equally important that Genomics England staff are shown to be experts. Such expertise would include demonstrating that they understand the field, know how to set up the data management processes and can set up robust approvals/ and ethics review processes. Clearly stating on Genomics England's website and in other communications who the staff are and their track history is essential according to participants.

> *"Qualifications and reasons as to why the people that are in charge of this study at Genomics England are there. They need to prove their, kind of, credentials as to why they're the people that are in charge of this, why we should trust them. If not qualifications then it's just credentials, if they've just got some kind of something in their past, or the experience doesn't necessarily have to be a physical, like, qualification, but just something that says, 'This is why I'm the guardian of this.'"* Participant, Southern online group

5. Highlight the safeguards in place

Being very clear about the safeguards in place for the principle of putting safety first was felt to be an essential demonstration of trustworthiness. When discussing this, participants emphasised the importance of:

- High levels of data security
- Informed consent with a clear process for withdrawing at any point
- Deidentification of the data – with explanations of what 'deidentification' means clearly expressed to all those who join the programme
- Establishing and communicating what the process is if something goes wrong
- Showing a track record in successfully managing the NGRL and previous projects which have involved researchers being granted access to sensitive genomic data.

6. Be accountable

Participants took Genomics England's role in designing, delivering, and managing the Newborn Genomes Programme very seriously. As such, they wanted to know that Genomics England will act responsibly and take responsibility for the decisions made now and in the future about the programme. If Genomics England demonstrates that they are accountable for what happens in the programme it will be trusted.

> *"Genomics England are ultimately going to be responsible for the people who are accessing the data, they'll do the vetting. They're making sure that the researchers are doing what they say they're going to do. Ultimately, somebody has to be responsible and therefore ultimately you have to put your trust in that person or organisation."* Participant, Liverpool in-person group

To be truly accountable, participants felt that Genomics England needed to be seen to have independent oversight. Many participants thought an oversight or an advisory committee should be in place for all of the Newborn Genomes Programme, including the discovery research element. They saw this as an overarching group which would sit above approvals and research ethics committees. Suggestions for who should be part of the committee included people with significant experience of genomic data collection and use. Participants also said they would also like to see public involvement in such a committee.

> *"We need to have the oversight committee from different places. Maybe we get somebody from universities on the committee as well, something from, like, the people who know about genomics. Maybe we could have someone from the public as well, or something. Something like that."* Participant, Southern online group

7. Participant feedback on the deliberation

Three participants who took part in the deliberation agreed to take on the role of citizen journalists and give feedback on the experience of the dialogue. These experiences are described in the participants' own words in the following section. Ending with the words of participants in the deliberation who committed significant time to the process is in a process which seek to gain a depth of understanding of views and opinions.

7.1 Adrian, northern England, participated online:

> *"The relevance of the information provided, and the quality of the facilitated group discussions enabled me and my fellow participants to have considered discussions around a variety of interesting issues such as consent, security and the ethics of data collection, storage and usage. We each had the chance to voice our opinions in a safe space where our contributions felt valued. Overall it was a very positive and rewarding experience with the added benefit of having contributed to something meaningful."*

In February 2023 I was fortunate to be asked to take part in a public deliberation concerning discovery research and the Newborn Genomes Programme. I had never heard of the Programme or even heard of Genomics England who have overall responsibility for it, so I was both intrigued and slightly nervous about what I was going to be involved in. I need not have been nervous. Even before the workshops started it was clear that this was being done in a sober and professional manner and we were given lots of relevant information to help prepare us for the series of workshops.

Our group met online through a series of four facilitated sessions where we considered a variety of interesting issues concerning the Newborn Genomes project. It was clear at the outset what each session was going to cover, how it was to be structured, timings and what the proposed outcomes were to be (as well as the overall outcomes across the four sessions). Areas that we considered included:

- Issues discovery research raises for consent – who gives it? In what circumstances could it be withdrawn and by whom?
- Ethical considerations raised by different discovery research scenarios
- Governance and oversight of discovery research – how would we have confidence that the data was being used appropriately? Who makes sure the people with the data are using it correctly?
- Security issues – how would we know the data is being held securely and is impervious to cyber-attacks? What happens if data is shared with third parties – especially those in foreign countries where UK law would not apply?

Each of the sessions was facilitated excellently and ran to time, but it was the smaller group discussions we had during each session which made this such a positive experience. These were small, facilitated groups of between 6-8 people where we were able to do a 'deep dive' around some of the issues identified above. The time spent on these enabled us to have a rich discussion and to consider issues in some depth before feeding back to the wider group. Smaller groups ensured that everyone had the chance to voice their opinion and discuss any points of disagreement or contention in a safe space where everyone was encouraged to contribute, and our perspectives were valued.

Each session lasted 2.5 hours. When combined with the background reading and contributions needed between each session, this meant quite a significant amount of time was spent on this topic. And yet, by the end of it, I was sorry that it was finishing and felt that I could have taken part in and contributed to further sessions. This was an interesting and fulfilling experience where there was a real sense that we were contributing to something meaningful and worthwhile. I can't wait to see the results of our discussions and the introduction of the Newborn Genomes Programme and to feel the satisfaction of knowing I contributed in my own small way to its introduction.

7.2 Amber, southern England, participated online:

> *"My group was made up of a variety of individuals, all from various backgrounds including some who had their own experiences with genetic diseases or disorders. Speaking with them about the topics of discussion underlined the great importance of discovery research, as it showed that this type of research has real-world impact for everyday people like you and me."*

Participating in this public deliberation on research with data from Genomics England's Newborn Genomes Programme was a thought provoking process. There were four workshops held across February, with some pre-workshop exercises to help put our participation into context. After being introduced to the timeline of the workshops and what each workshop would cover, we were split into smaller groups of about six or seven for more focused discussions.

Before beginning the workshops, I had very little knowledge of studies that are being undertaken to better understand genetic diseases. I have been fortunate enough that I have never suffered with any genetic diseases, nor known anyone who does. I knew the importance of discovery research but was very sheltered from the huge impact a breakthrough in that field could have.

My group was made up of a variety of individuals, all from various backgrounds including some who had their own experiences with genetic diseases or disorders. One individual had ADHD, another had a child with a genetic disorder. Speaking with

them about the topics of discussion underlined the great importance of discovery research, as it showed that this type of research has real-world impact for everyday people like you and me.

I found the research scenarios very useful to the discussion. These scenarios framed the impact of researchers having access to the data held in the National Genomic Research Library (NGRL) in a context that I could understand. From the scenarios and consequent discussions with my group, I considered how discovery research could do much more than improve understanding of where genetic diseases come from. It could help to diagnose children sooner, it could develop more effective drugs to treat children, it could allow the NHS to tailor their support to affected families in more effective ways.

Something that took my interest was how this kind of research could bring about wider social change. One specific scenario that was given to us was about research into obesity, and how pairing the data from the Newborn Genomes Programme with other national data sets could determine how much of childhood obesity is genetic, and how much is environmental. Researchers could publish their findings alongside advisories for parents, schools and governments about what could be done to reduce childhood obesity on an environmental level.

There was a lot of concern around data security. Everyone in my group collectively agreed that consent, the ability to withdraw consent, and patient confidentiality was at the core of being able to carry out this kind of research ethically.

7.3 Gbemi, London, participated in person:

> *"In some ways it feels like the starting point, but it seemed clear that research like this could lead to more help or interventions in the future... Overall, I think the more researchers can work with openness from the beginning, the more transparency there is, the more likely people will be to open up themselves up to this kind of research."*

I was interested to take part in these workshops because I wanted to know more about genes and their impact on different kinds of health conditions. I have sickle-cell disease myself, so from a personal perspective I am particularly interested in finding out more about rare disorders and how earlier diagnosis can help with earlier treatment.

The workshops I took part in were held on two Saturdays, three weeks apart. We were provided with videos and online activities before coming to the first workshop. This helped me to understand more about the topic we were exploring and gave me the confidence to fully participate in the group discussions in person. At the workshop, the facilitators really got us thinking for ourselves, they didn't just feed us information but got our brains to work for it.

The sessions were quite long and asked a lot of us, but I don't think a topic like this could have been covered in less time. By providing us with a timetable and breaks it was clear the organisers had us in mind, not just what they needed from the day. There were lots of questions about the topic, but organisers were on hand to answers these and when questions occurred to me after the first workshop, it was good to know I would have an opportunity to ask these either at the next workshop or in the online space.

It was especially interesting for me to learn more about the different rare conditions that researchers could study using data from the Newborn Genomes Programme. It opened my eyes to the ways science might lead to possible innovations. In some ways it feels like the starting point, but it seemed clear that research like this could lead to more help or interventions in the future. For me, the speed of diagnosis is important, as this could lead to earlier interventions and treatment, whereas having to wait longer for a diagnosis could lead to more difficulties or complications.

There are risks attached to innovation and the more innovation there is the more the important the checks and balances need to be. However as long as there are consequences for the misuse of data I think this can be managed. For instance, breaches of privacy should lead to legal consequences and possibly elimination from carrying out research in the future.

Overall, I think the more researchers can work with openness from the beginning, the more transparency there is, the more likely people will be to open up themselves up to this kind of research. If there is less transparency, people will feel more resistance and will be more likely to have doubts about the work and distance themselves from it. Transparency will help to build trust and people will feel more comfortable sharing their information and keeping in contact with the programme. Following our discussions, my hopes for the Newborn Genomes Programme are for it to remain open and transparent, continue to run workshops and keep people informed, so that people can learn about their work, and they can learn from us.

7.4 Acknowledgements

Hopkins Van Mil is enormously grateful to participants from across the UK who took part in this public deliberation process. Their commitment to the workshops was sincere and thoughtful. Their lively and passionate contributions on a complex subject meant we have been able to share their views on discovery research within the Newborn Genomes Programme in a way which will support the programme to achieve its aims.

Particular thanks are due to Kate Harvey, Amanda Pichini, Alice Tuff-Lacey and Natalie Banner from Genomics England who gave significant time to guiding this process. We are also most grateful to them for presenting to our dialogue participants and to providing key information on the study and discovery research scenarios without which the discovery research aim could not have been considered by participants.

Many thanks too to Professor Paul Gissen, Clinical Professor in Paediatric Metabolic

Medicine at UCL Great Ormond Street Institute of Child Health; Professor Matt Hurles, Head of Human Genetics at Wellcome Sanger Institute; Professor Neena Modi, Professor of Neonatal Medicine at Imperial College London and Consultant in Neonatal Medicine at Chelsea and Westminster NHS Foundation Trust; Professor Sir Munir Pirmohamed, Professor of Pharmacology and Therapeutics at University of Liverpool; Dr Nadeem Sarwar, Global Head of Genomic Strategies and Digital Strategies, at Eisai; Professor Reecha Sofat, Professor of Pharmacology and Therapeutics at University of Liverpool. Each of these speakers gave up their time to give fascinating insight to participants to inform their deliberations. We are most grateful.

Hopkins Van Mil 27th April 2023

Appendix A – Methodology

Genomics England commissioned Hopkins Van Mil (HVM) to begin work on this public deliberation on research access to Newborn Genomes Programme data in January 2023. It was necessary for the project to be completed within a three month period. The project's initial phase, consisting of scoping and design, was completed by the end of January.

HVM project team members attended induction meetings, presentations and Q&A sessions with wider members of the Newborn Genomes Programme ('the Programme') team during this time to quickly get up to speed with the Programme. These sessions and ongoing communication with Genomics England including weekly project management meetings helped to identify essential reading materials which informed HVM's desk research on the project. Awareness and understanding of the topic was also developed through desk research HVM had previously conducted during its work on a public dialogue on Implications of Whole Genome Sequencing for Newborn Screening for Genomics England, the UK National Screening Committee and Sciencewise in 2021[7]. Senior members of the HVM project team working on the public deliberation had experience from this dialogue which directly informed their work on the current project.

A deliberative process

Public deliberation was intentionally chosen as the methodology to bring a depth of understanding on public attitudes to discover research in this context. Public deliberation is not a 'we tell you this and you tell us what you think about it' information exchange. Deliberation works when participants interact on a level playing field with specialists: academics, researchers, scientists, and policy makers. In this deliberation this included clinicians, scientists working in academic and industry settings and an ethicist. This specialist evidence is then viewed through the lens of participants' lived experience which leads to rich and powerful insights.

In a public deliberation citizens come together with sufficient time to reflect, to:

- Learn about the issue
- Talk with, not past, each other
- Consider diverse points of view
- Discover key tensions and values
- Spark new ideas

This leads to an understanding of what people value, what they see as benefits and harms, their trade-offs and redlines and, in this case, highlights areas of importance when assessing research access to newborns' genomic data.

Dialogue uses expert facilitators. In this case each online workshop and in-person workshop had 4 facilitators, including a lead facilitator. We used a consistent group of facilitators and this number of team members, plus technical support, allowed us to have small groups of no more than 7 participants. Such ratios allow trust to build

[7] The implications of whole genome sequencing (wgs) for newborn screening, July 2021, Hopkins Van Mil with Genomics England, the UK National Screening Committee and Sciencewise.

and a greater depth of exploration of the issues. Facilitators followed workshop process plans designed in discussion with the Project Team.

Recruitment

101 people took part in the deliberation. Each person was recruited into one of four workshop groups depending on their location: Liverpool (in-person), London (in-person), Northern England (online), Southern England (online). With the exception of the Liverpool and London groups, for which all participants were recruited from the city and its surrounding areas, all other participants were recruitment from across England drawing from urban, rural and suburban communities. Recruitment into the Northern England group included the following regions: North East, North West, Yorkshire and Humber, East Midlands, West Midlands. Recruitment into the Southern England group included: East of England, London, South East, South West.

Overall the group was a broadly reflective sample which was weighted to increase, in relation to current census data, the number of people drawn from communities experiencing racial inequalities, disabled people, those with long-term health conditions and people with and parents of children with genetic conditions.

A specification and screener were used to ensure that as far as possible, participants reflected the demographics of the population of England, sampling for age, ethnicity, gender, life stage, disabilities and socio-economic group beyond the weighting described above. We excluded those who had taken part in deliberative type activities in the previous 12 months, as well as anyone whose profession closely overlapped with the focus of the deliberation. Participants in the deliberation were given a cash honorarium to recognise the time committed. This reflects standard practice in Sciencewise public dialogues and means people are not excluded because of their financial circumstances.

Potential participants also answered behaviour and attitudinal questions to explore their current relationship to the topic and perceptions of health research. We asked:

- Are you already part of a health research database – e.g. a birth cohort, biobank?
    - Yes
    - No
    - Not sure/prefer not to say

- ‘I have heard of the 100,000 Genomes Project’
    - Yes
    - No

- On a scale of 1-5 (where 1=not at all hopeful and 5=very hopeful), how hopeful are you about using research data to make improvements in healthcare?

- On a scale of 1-5 (where 1=no trust at all and 5=a great deal of trust), how much trust do you have in health research conducted by the NHS?

These questions were used to understand the spread of views on data collection and

use rather than as a reason to exclude or include participants in the dialogue.

Workshop process

The dialogue took the form of two full day workshops (on 4th and 25th February) for participants in London and Liverpool who took part in-person. For those who took part online (groups from Northern and Southern England) the workshops took place over four week-day evenings from 7th to 23rd February. Table 1 sets out the discussion points for each round of workshops.

Table 1

Round one	Round two
London/Liverpool: Workshop 1, Saturday 4th February (10:00am-4:00pm) Northern/Southern England: Workshops 1 & 2, Tuesday 7th and Wednesday 8th February (6:00pm-8:30pm both sessions)	London/Liverpool: Workshop 2, Saturday 25th February (10:00am-4:00pm) Northern/Southern England: Workshops 3 & 4, Tuesday 21st and Thursday 23rd February (6:00pm-8:30pm both sessions)
• Introductions to the team and settling into the topic • An introductory presentation from Genomics England on the Newborn Genomes Programme • Introduction to and presentations on discovery research • Initial exploration of the things people consider important when thinking about researcher access to Newborns' genomic data • What, if anything, is significant about the fact that it is newborns' data? • About data's role in the NHS and research • Thoughts about different discovery research use cases and their levels of acceptability • 2/3 discovery research scenarios: epilepsy, obesity, attention deficit hyperactivity disorder (ADHD)	• Re-introductions to the team and focus and purpose of the deliberation • Discovery research scenario: Rett syndrome • Further presentations on discovery research • Presentation on ethical issues around discovery research and the Newborn Genomes Programme • Exploration of trust and trustworthiness in the context of research access to genomic data and linkage with other datasets • Reflections on data linkage, including with maternal health data • Discovery research scenario: Condition X • Final considerations about transparency, governance and communications

It is an essential part of public deliberation that participants interact with specialists in

the field. This exposes participants to a range of examples and perspectives on the issues. We therefore worked with a range of specialists to provide expertise in the form of presentations, answering questions, responding to comments with additional evidence, information, and a range of opinions. Table 2 provides a list of the speakers involved across the deliberation.

Table 2

Workshop speakers

Presentations were primarily delivered live either in-person (London/Liverpool) or on Zoom (Northern/Southern England), with recordings uploaded to the online space Recollective for all participants to watch.

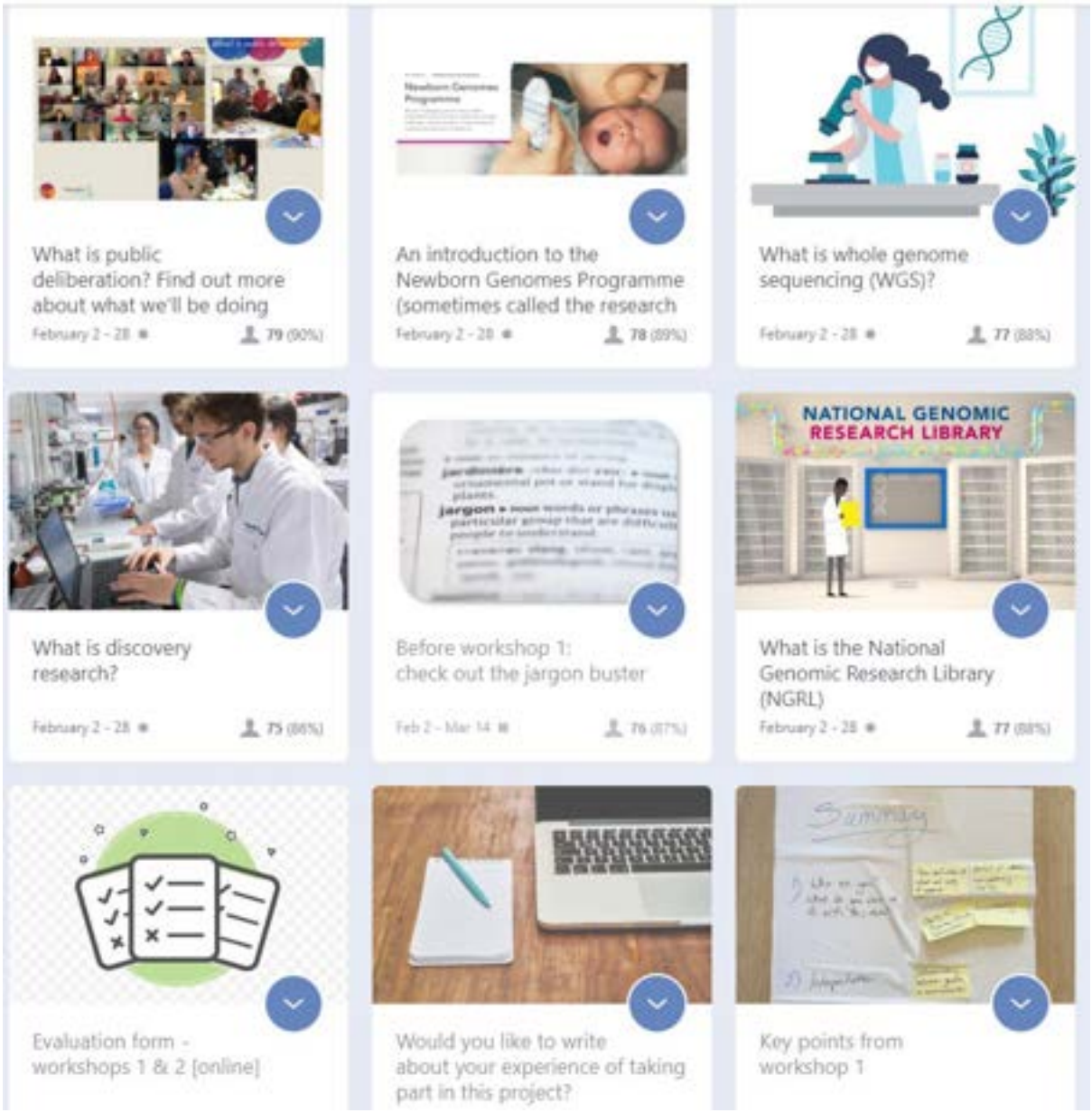
Dr Natalie Banner	Director of Ethics at Genomics England
Professor Paul Gissen	Clinical Professor in Paediatric Metabolic Medicine at UCL Great Ormond Street Institute of Child Health
Kate Harvey	Engagement Manager, Genomics England
Professor Matt Hurles	Head of Human Genetics at Wellcome Sanger Institute
Professor Neena Modi	Professor of Neonatal Medicine at Imperial College London and Consultant in Neonatal Medicine at Chelsea and Westminster NHS Foundation Trust
Professor Sir Munir Pirmohamed	Professor of Pharmacology and Therapeutics at University of Liverpool
Dr Nadeem Sarwar	Global Head of Genomic Strategies and Digital Strategies, at Eisai
Professor Reecha Sofat	Professor of Pharmacology and Therapeutics at University of Liverpool

The online space

We asked participants to spend time before the deliberation, and in between each workshop, in an online space called Recollective which was tailored to this project. We asked participants to come to the first workshop having watched a video explaining the deliberative process. They were also asked to watch introductions to:

- The Newborn Genomes Programme
- Whole Genome Sequencing
- Discovery research
- The NGRL

This contextual information meant that they could come to the first workshop with a clear idea of the subject under discussion and the process for discussing it.



An image from the online space summarising some pre-workshop activities

Participants were then asked to use the online space to review the points made by each small group in the discussion and to rewatch recordings of the presentations made. We also asked participants to do some thinking around our deliberation topics before coming to workshops, for example, they had an activity on trust and several activities about discovery research to think through.

In this space we also reshared the scenarios used in the deliberation and asked people to comment further on them in that space. Answers to any questions that weren't fully answered in the workshops were also shared there.

Analysis & reporting

Essential to this process is the capture of views to make sure the findings fully reflect the participant voice. All data collection remained robust throughout, both in online

and face-to-face workshop settings. Each facilitator recorded their own small group discussions, and the plenary discussions were audio recorded, with the chat also being saved (online workshops only); the latter providing a useful source of real-time commentary in the participants' own words.

At each workshop facilitators took visible notes using flip charts (in-person) or by sharing their screens whilst typing (online). In both settings participants could amend what was written, review what they had discussed and prioritise key points made as required. As such, these were not part of the data capture process but were useful in understanding the points on which participants had placed particular emphasis and have been drawn on to some extent in our analysis. In addition, comments made in the dedicated online space were captured and analysed.

All the qualitative data was captured on the coding and analysis tool NVivo. Facilitators met in a team analysis workshop to agree an initial coding framework. A discussion was held with the Genomics England project team to agree the report structure. The HVM team of analysts and report writers worked together to create this report, which was reviewed in draft by Genomics England before being confirmed in its final version.

Appendix B – Process materials

The full set of process materials is available from Hopkins Van Mil on request. Below we share a sample of the process plans used for the in-person workshops. These were adapted for the online workshops, so that participants had a parity of experience in whichever format they worked with the HVM team.

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
8:30-9:45	Set-up	Room set up – cabaret style: • 4 tables for 9/10 • 4 x flip charts • 4 x facilitation kits • Welcome table – registration list, research sign in, photography sign-in • Participant materials • Projector, speaker box, laptop loaded with films/ PPs and with access to Mentimeter • Menti QR code/ number code printed on each table	HVM team		Project team set up and ready
9:30-9:45	Speaker briefing	LF to speak to any observers/ speakers present	LF	Speaker guidelines	Speakers ready to join in
9:45-10:00	Participant Check-in	Event support +1 at welcome table • Greet participants • Give them welcome pack and bag • Signpost loos, refreshments, their table • Answer any questions	Event support + facilitator		Participants set up and ready

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes

10:00-10:25 (25 mins)
 	
10:00-10:10 (10 mins)
	Introduction & workshop purpose	Warm welcome to the first of two workshops, setting the tone for the session. Delighted to welcome you here for the first of two workshops, both held here.
		
		1. Housekeeping
		2. LF intro. HVM team stand up and introduce themselves
		3. Any observers/ speakers present introduce themselves
		
		Then LF:
		• Refers to using Recollective in between workshops
		• Shares today's programme – where we will be setting the context of our deliberations and thinking broadly about discovery research,
		• Shares the points to help the discussion
		• Presents the purpose
		• Explain the handbook
		• Explain how the small groups work
		• How we record the session:
		  ○ Audio recorders on each table
		  ○ Flip notes
		  ○ Post-its
		  ○ Any other thoughts cards
		• We need to record what you have said so that we can produce a report for Genomics England which clearly reflects your views. We are interested in what you say, not who has said what. Your name will never be linked back to any comment you have made in these discussions in our reporting. Our transcripts
			HVM
			
			LF using HVM slides
				PP Purpose & Agenda Slide
				
				Intro PP
				
				Menti on the screen
					People are clear:
					Who is in the room and why;
					who they will be working with
					What we will be doing together
					What the scope of the discussion is
					How we will work together and how the findings from the deliberation will be used

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
10:10-10:15 (5 mins) 10:15-10:25 (10 mins)	Menti.com	are anonymised so no names appear in the material that we analyse to produce our report. More information is on the data protection information in your pack. • Shows how this workshop fits with everything else that's happening • What will happen as a result of our work together – the findings will be written up in a report. Genomics England will use these findings to inform the design and development of the Newborn Genomes Programme's research study. End with Fergus Walsh BBC film clip here. (make sure no recording) **Participants asked to get menti.com on their phones:** Or ask their facilitator/ event support to do it for them from their phone if they don't have access/ would rather not. QM1: Share one quick thing about yourself QM2: When I say 'health research' what comes to your mind? QM3: To what extent have you thought about health data being used for research and innovation in healthcare as well as for individual care? • I have never thought about it • I have thought about it occasionally • I have thought about it regularly •			
10:25-10:45 (20 mins)	Getting in the zone	Whole room working together guided by LF.	LF	Pics: Newborn	Participants get to briefly

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
10:25-10:35 (10 mins) 10:35-10:45 (10 mins)		Each person has a picture card on the table in front of them. There are no right or wrong answers here, we're just getting to know each other. Please stand up and walk around the room holding up your card. **1. Picture match:** Find the person who has the same card as you. Briefly introduce yourself to the person you've found. Talk together about what the picture means to you and why, mention any stories from your own life if you'd like to. When the bell rings: **2. Go back to your original table** **RECORDER ON** Go round the table. Each person says hello to the group: Your name, and 2 highlights from the conversations you've just had. What the pictures meant to you and those you spoke to. **RECORDER OFF**		health check Health research Imperial research A visualisation of health data Ambulance service Maternity unit Heel prick test	meet everyone in the room before focusing in on small group discussions.
10:45-11:15 (30 mins) 10:45-10:55 (10 mins)		Come back together as a whole group for two presentations. **1. GeL – introduction to the research study** A recorded film from Genomics England As you are listening note down any questions you have on post-it	LF ES Speakers	LF PP Film downloaded	Gaining understanding of the programme, discovery

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
10:55-11:00 (5 mins) 11:00-11:10 (10 mins)		notes. We'll gather these up and get answers for you. Short intro from LF **2. What is discovery research?** Definition on screen and on the wall (and already shared on Recollective): Genomics England have asked us to consider 'discovery research' within the Newborn Genomes Study with you. By this they mean health research which is exploratory. In time these explorations could lead to new knowledge, insights and contribute to future advances in healthcare and treatment; bringing positive changes for people's care. **Some points to think about:** • Researchers will be able to look at research questions that go beyond the 200 conditions the Newborn Genomes Programme will be testing babies for. • These research questions could be very broad. • It could even involve accessing data from the newborns' participants solely for the purpose of them acting as 'controls' for a research question involving other participants from our other cohorts. • Just because something is researched in the NGRL doesn't mean each person gets individual feedback of any research findings.		Ipad & tripod/ voice recorder Speaker PP as needed	research and how it relates to clinician's work. Initial questions gathered and answered where possible.

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
11:10-11:20 (10 mins)		A reminder from what you have reviewed in the online space: The newborns who will be involved in our study will be largely healthy. We expect that, out of the 100,000 babies who take part, only 500-1000 will receive a result that indicates they have a rare genetic condition. **Presentations to be recorded by Event Support** **3. Presentation** As our speaker is presenting take a post-it and write down any questions you have. How research access for discovery research works in my field: **Liverpool**: **Professor Sir Munir Pirmohamed,** Professor of Pharmacology and Therapeutics, University of Liverpool **London**: **Professor Neena Modi**, Professor of Neonatal Medicine at Imperial College London and Consultant in Neonatal Medicine at Chelsea and Westminster NHS Foundation Trust **RECORDER ON** Q&A facilitated by LF **RECORDER OFF**			
11:20-11:35 (15 mins)	**Break – refreshments available**				

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
11:35-12:15 (40 mins) 11:35-11:50 (15 mins) 11:50-12:20 (30 mins)	A discussion on discovery research	Back at tables to work in small groups. Your first thoughts at this point in the deliberation about data access for discovery research: **Q1: What do you think it is important to consider when thinking about researchers having access to newborn genomes data?** *Remember what you learnt on the online space – that the data is held in the NGRL. This is a reference not a lending library.* • Work in 2s/3s. • Use post-its • Given what you've seen in the online space and heard so far this morning - think about as many things as possible that should be considered when researchers have access to newborns genome data • You might want to think about this from different perspec-tives e.g. the newborn, the parents, the researcher, Ge-nomics England • Write all these things down on the post-its • One post-it per thing. **RECORDER ON** Let's discuss what you've found together. Each 2/3 to share their post-its, Facilitator to collate these on a flip	Fs	Post-its Pens Flip paper	Understandi ng how participants see the scope of this discussion shaping up. Getting their front of mind thoughts about some of the specifics of newborns' genomic data. We are looking at this in more detail this afternoon when we will think about: - What data? - What kind of research?

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
		sheet – if the following headings work for each sheet, use them – or create other collation headings if more appropriate: • Opportunities/ hopes – for newborns, parents, researchers, Genomics England, Society • Challenges/ concerns – for newborns, parents, researchers, Genomics England, Society Prompts for initial discussion: • Why was this a consideration? • What specifically were you concerned about here? • What specifically did you see as an opportunity here? • Can you give me an example of that? As a result of this discussion we have a long list of considerations when we think about researchers having access to data from the newborn genomes programme. Looking at this long list: **Q2: What from our list of considerations indicates specific points about the use of *newborns'* genomic data for discovery research?** Prompts – to be used as necessary, but see what emerges spontaneously first: • To what extent are there unique issues/ sensitivities when we think about newborns' data? E.g. a newborn can't consent for itself. • That most of the babies that take part will be healthy/ won't have a genetic condition.			- Where researchers are from?

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
		• Facilitator to make flip chart notes of key points – so that they can be picked up later/ explored further in the process. This is the first of our discussions on this. We'll pick up these points again as continue to work together. **RECORDER OFF** Group to turn back to face the central screen			
12:20-12:35 (15 mins) 12:20-12:30 (10 mins)	The 'data in the NHS and the data to research' journey	LF to play two animations: **1. UPD: Data saves lives** stop at 2:07 https://youtu.be/fJ2hyXCOOyQ 'and ensure data is shared responsibly.' **2. The 100,000** genomes project film clip: . Starting at 1:48, 'before we let research scientists look at your data'. But first explain this is an example of the foundational project for Genomics England: • When it refers to 'your data' in our case we are talking about newborns' data • It refers to 'the Genomics England Data Centre', this is now the 'National Genomic Research Library (NGRL)' you've already heard about. **Participants asked to get menti.com on their phones:** Or ask their facilitator/ event support to do it for them from their phone if they don't have access/ would rather not.			

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
12:30-12:40 (10 mins)	Menti.com	QM3: Share one question you have about what you have read/ heard about the use of data in the NGRL. QM4: What comes to your mind when you think about newborns' data being accessed by researchers?			
12:40-1:05 (30 mins) 12:40-12:55 (15 mins)	Exploration of the data journey	Back in small groups We are now thinking specifically about the Newborn Genomes Programme and the data it will use. The programme will collect data on 100,000, largely healthy, newborn babies. We are going to explore how this data might be accessed for discovery research. Work in 2/3s – use post-its, one use per post-it. **Q3: What are all the discovery research uses that you can think of using data from the Newborn Genome Programme?** Prompts – to be used as necessary, but see what emerges spontaneously first – note - some uses are likely to overlap: • Develop understanding about future health needs across the population resourcing needs • Gain new scientific knowledge about diseases and condi-tions • Find new ways of supporting people with genetic conditions • How to improve/ repurpose current therapies • How to develop new diagnostics/ treatments. Come back together to discuss those uses.	Fs	Post-its Flip sheet for post-it collation and	Thinking through discovery research uses

Workshop 1, round 1 – in person					
Time	**Agenda**	**Process**	**Who?**	**Process Tools**	**Expected Outcomes**
12:55-1:10 (15 mins)		1. Facilitator to ask people to share their post-its, creating a flip sheet or two with all the uses. Then: **Q4: Thinking through these uses, and what you've learnt so far – what uses, if any, feel acceptable? What uses, if any feel unacceptable, what uses are you not sure about?** **RECORDER ON** Group to discuss: • What feels acceptable and why? • What feels unacceptable and why? • What are you not sure about and why? **RECORDER OFF**		notes. Highlighting any priority points.	
1:10-1:50 (40 mins)	**Lunch**				
1:50-3:00 (60 mins) 1:50-2:05 (15 mins)	Exploration of scenarios 1. Epilepsy	Back in small groups **RECORDER ON** **Epilepsy discovery research** – the detail is in your handbook on page 13. Facilitator to read through the scenario with their group **Q5: What is your view on this data research access request?** • What feels good about it - why?	Fs	Facilitator to note key points on the flip chart. Highlighting any priority points.	Getting into the detail of discovery research – what's good/ what's challenging and what they expect/

Workshop 1, round 1 – in person					
Time	**Agenda**	**Process**	**Who?**	**Process Tools**	**Expected Outcomes**
		• What feels challenging about it - why? **Q6: What would you expect from the researcher using newborns' genome data for research as this?** • What assurances would you need about using the data for this purpose? • What would you hope for and why? E.g. from Genomics England, from the researcher			hope
2:05-2:20 (15 mins)	2. Obesity	**Obesity discovery research** – the detail is in your handbook on page 14. Facilitator to read through the scenario with their group **Q7: What is your view on this data research access request?** • What feels good about it - why? • What feels challenging about it - why? **Q8: What would you expect from the researcher using newborns' genome data for research as this?** • What assurances would you need about using the data for this purpose? • What would you hope for and why? E.g. from Genomics England, from the researcher			
2:20-2:35 (15 mins)					
2:35-2:50 (15 mins)					
2:50-3:05 (15 mins)	**Break – refreshments available**				
3:05-3:45 (30 mins)	Scenarios 3. ADHD	**Attention deficit hyperactivity disorder (ADHD)** – the detail is in your handbook on page 15. Facilitator to read through the scenario with their group		Facilitator to note key points on the	Getting into the detail of discovery

Workshop 1, round 1 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
3:05-3:20 (15 mins) 3:20-3:35 (15 mins) 3:35-3:45 (10 mins)		**Q9: What is your view on this data research access request?** • What feels good about it - why? • What feels challenging about it - why? **Q10: What would you expect from the researcher using newborns' genome data for research as this?** • What assurances would you need about using the data for this purpose? • What would you hope for and why? E.g. from Genomics England, from the researcher **Q11: What 3 points summarise the discussions we've had on these three scenarios this afternoon?** Facilitator to create summary sheet on the flip chart. These will be photographed and put up on Recollective.		flip chart. Highlighting any priority points. Flip chart: Summary points: 1, 2, 3	research – what's good/ what's challenging and what they expect/ hope
3:45-4:00 (15 mins)	Menti.com Wrap up and close	Back to working as a whole group QM5: One thing that you have learnt or has particularly interested you from today's discussions QM6: One thing you hope for from this public deliberation Share the evaluation form Reminder to come back on 25th February, same time same place. Keep us in your minds with the activities that will go up on the online space between now and then.		Menti	Summing up and clarity on next steps.

Workshop 1, round 1 – in person					
Time	**Agenda**	**Process**	**Who?**	**Process Tools**	**Expected Outcomes**
		Thanks for being with us today			
Online space	Recollective	• Review the key point summaries from each group • Review the presentations and videos again (if you'd like to) • Look at the scenario on Rett syndrome – what are your views on this data access request? • Think about a situation where data about you/ your children/ other family members is being collected and used for research – what makes such system trustworthy?			

Workshop 2, round 2 – in person					
Time	**Agenda**	**Process**	**Who?**	**Process Tools**	**Expected Outcomes**

8:30-9:45	Set-up	Room set up – cabaret style:
		• 4 tables for 6/7
		• 4 x flip charts
		• 4 x facilitation kits
		• Wifi sign-in
		• Welcome table – registration list, research sign in, photography sign-in
		• Participant materials:
		  ○ Scenarios
		  ○ name badge
		• Projector, speaker box, laptop loaded with films/ PPs and with access to Mentimeter
		• Menti QR code/ number code printed on each table
			HVM team		Project team set up and ready

9:30-9:45	Speaker briefing	LF to speak to any observers/ speakers present	LF	Speaker guidelines	Speakers ready to join

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
					in
9:45-10:00	Participant Check-in	Event support +1 at welcome table • Greet participants • Replacement handbook if they haven't brought theirs • Copies of the 2 scenarios being used if they have forgotten theirs • Re-sign the audio/ video/ photo film form • Signpost loos, refreshments, their table • Answer any questions	Event support + facilitator		Participants set up and ready
10:00-10:25 (25 mins) 10:00-10:15 (15 mins)	Introduction & workshop purpose	Warm welcome to the second of two workshops, setting the tone for the session. 1. Housekeeping 2. LF intro. HVM team stand up and introduce themselves 3. Any observers/ speakers present introduce themselves Then LF: • Refers to using Recollective in between workshops • Shares today's programme – where we will be setting the context of our deliberations and thinking broadly about discovery research, • Shares the points to help the discussion • Presents the purpose • Runs through what we've discussed before including scenarios • Runs through the discovery research model	HVM LF using HVM slides	PP Purpose & Agenda Slide Intro PP	People are clear: Who is in the room and why; who they will be working with What we will be doing together What we've done so far and some information to inform the deliberations

Workshop 2, round 2 – in person					
Time	**Agenda**	**Process**	**Who?**	**Process Tools**	**Expected Outcomes**
10:15-10:25 (10 mins)	Menti.com	• Reminder of recording and of how the report we are working on will be used. **Participants asked to get menti.com on their phones:** Or ask their facilitator/ event support to do it for them from their phone if they don't have access/ would rather not. QM1: One thing you remember from our last workshop? QM2: One concern you have about research access to newborn genome data? QM3: One hope you have about research access to newborn genome data? •		Menti on the screen	
10:25	**Work in small groups**				
10:25-10:50 (25 mins) 10:25-10:35 (10 mins) 10:30-10:40 (10 mins)	Getting straight back into our work	Fs reminder of the recording. **RECORDER ON** Go round the table – quick introductions • Reintroduce yourself to the group **Q1: On the menti you shared one thing you remember from our last workshop – why did this come to your mind?** Talk in 2s/3s – think about the Rett Syndrome scenario you looked at in the online space over the last week.	Fs	Handbooks Additional copies of the scenarios as needed	Reminder of who is on the table Getting back into the space. Gathering thoughts on a scenario people have

Workshop 2, round 2 – in person					
Time	**Agenda**	**Process**	**Who?**	**Process Tools**	**Expected Outcomes**
10:40-10:50 (10 mins)		**Q2: What is your view on this data research access request?** • Note down on post-it's the main things that you thought about this research access request. • One thought per post-it • Come back together as a group. Gather up the post-its and collate them on the flip sheet Discuss: • What feels good about it - why? • What feels challenging about it - why? • What would you expect from the researcher using new-borns' genome data for research as this? **Facilitator note:** no need to cover this in depth, we have people's responses to the scenario on Recollective. **RECORDER OFF**		Post-its Sharpies Flip sheets	already considered. Reviewing examples of discovery research and their implications.
10:50	**Come back together as one group**				
10:50-11:10 (20 mins) 10:50-11:00 (10 mins)	A further example to inform discussions	Come back together as a whole group for a presentation. As our speaker is presenting note down any questions you have on post-it notes: How research access for discovery research works in my field:	LF Speakers	LF PP Ipad & tripod/ voice	Gaining understanding of the programme, discovery research and how it

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
11:00-11:10 (10 mins)		**Liverpool**: **Professor Reecha Sofat,** Professor of Pharmacology and Therapeutics, University of Liverpool **London**: **Professor Nadeem Sarwar,** Global Head, Genomic Strategies & Global Head, Digital Strategies at Eisai. He works on the application of genomic and digital technologies in delivering innovation. He relocated back to the UK for this role because of the internationally-leading position of the UK genomics, data sciences and digital technologies. Prior to working in the pharmaceutical sector, Nadeem he was at the School of Clinical Medicine, University of Cambridge. Q&A for clarification/ understanding **RECORDER ON** Q&A facilitated by LF **RECORDER OFF**		recorder Speaker PP as needed	relates to clinician's work. Initial questions gathered and answered where possible.
11:10-11:25 (15 mins)	**Break – refreshments available**				
11:25-11:45 (20 mins) 11:25-11:35 (10 mins)	Ethical implications	LF introduce Natalie Banner (in person in London/ on film in Liverpool) **Dr Natalie Banner**, Director of Ethics at Genomics England. Natalie is responsible for enabling Genomics England to navigate the complex ethical challenges in advancing genomic medicine and research, and ensuring the organisation is a good steward of	NB	PP as needed Flips & post-its	Highlighting the ethical dimensions to the discussion on research access

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
11:35-11:45 (10 mins)	Menti.com	research participants' data. The ethical implications of research access to newborn genomes data. As Natalie is speaking please write any questions you have on a post-it. QM4: Share one question you have about what Natalie has said about the ethics of research access to newborn genomes data. **Liverpool** - We'll ask Natalie to review these – explain we'll get responses over lunch and share them with you by the end of the morning. **London** – live Q&A using the questions that have come up on menti		Menti.com	
11:45	**Back to small groups**				
11:45-1:00 (75 mins) 11:45-12:10 (25 mins)	Trust in data access/ Genomics England/ NGRL	Exercise to explore what we mean by 'trust', 'trusted' and 'trustworthy'. Contextual exercise drawing on scenarios outside this data access to the NGRL example and consider trustworthiness. Participants own lived experience of a system which is trustworthy. **RECORDER ON** In small groups:		Flips Post its	An understanding of the principles behind 'trust' and what it means to be trustworthy.

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
12:10-12:25 (15 mins) 12:25-12:40 (15 mins)		Go round the group, share: • Your example of trustworthiness • Why this demonstrated trust, what was the factor that made it trustworthy? Facilitator to create a flip chart sheet collating what was trusted and then a summary list of why, testing this with the group to check understanding. We now have a list of things which indicate why you had trust in the examples you shared. **Q4: What does what we have on this list tell us about what might be relevant in terms of trusted research access?** • What principles apply to trusted discovery research? • What does trust look like in relation to researchers access-ing newborn genome data? **Q5: To what extent is trust impacted when data linkages are made?** Reminder: we are still talking about de-identified data e.g. not data linked to an individual health record. Examples might include: - other data held on the newborns within the Newborn Genomes Programme e.g. on health - comparing healthy newborn genomic data with data in other parts of the NGRL such as from the 100,000 genomes project (on people with rare genetic conditions/ cancer)			

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
12:40-12:55 (15 mins) 12:55-1:05 (10 mins)		- linking de-identified data from the Newborn Genome Programme with nationally reported data on height, weight, education. • Positive impacts on trust • Negative impacts on trust • What would harm/ damage the trust built? **Q6: What should Genomics England do to demonstrate they are a trusted guardian for newborn genome data?** *Prompt to be used only if necessary* • What would ethical behaviour look like? E.g being held accountable/ involving people/ consent processes *Facilitator to create a summary sheet:* (If time) 3 key points on trust when considering research access for discovery research. Facilitator to create a summary flip sheet. **RECORDER OFF**			
1:05-1:45 (40 mins)	**Lunch**				
1:45-2:05 (20 mins)	Linking maternal data sets –	Go to small groups **RECORDER ON**	LF	Flips with 3 summary points from	Key points emerge on this issue

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
	a specific example.	**Q7: What do you feel about linking newborn genome data to maternal health data in the NGRL?** Assume this can happen – detailed being worked out – what do you think? • Acceptable/ or not? • Views on whether it changes the nature of the Newborn Genomes Programme to also collect maternal data? • To what extent does linking de-identified data on mothers and babies pose any concerns?		each group	which is a live one for GeL at this point.
2:05-2:45 (35 mins) 2:05-2:25 (20 mins)	Final exploration of scenarios Condition X	Stay in small groups – new topic area **RECORDER ON** **Condition X discovery research** – the detail is in your handbook on page 19. Facilitator to read through the scenario with their group Given everything we have discussed on ethics and trust. **Q8: What is your view on recontacting families in this context?** • To what extent is it acceptable to recontact families in this example? • Who should be making contact – why? • What, if anything, feels challenging about it - why?	Fs	Facilitator to note key points on the flip chart. Highlighting any priority points.	Focusing in on recontacting – what's good/ what's challenging and what they expect/ hope

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
2:25-2:45 (20 mins)		**Q9: What would you expect from the researcher using newborns' genome data for research such as this?** • What assurances would you need about using the data for this purpose? • What would you hope for and why? E.g. from Genomics England, from the researcher			
2:45-3:00 (15 mins)	**Break – refreshments available**				
3:00-3:45 (45 mins) 3:00-3:25 (25 mins) 3:25-3:40	Transparency, governance	Given all you have heard in both our workshops: **Q10: What would you expect:** • **expectant and new parents** • **and wider society** **to know about research access to Newborn Genomes Programme Data?** • What should we know, what would we want to know, what would we hope to know? • What would make the processes for research access trans-parent and clear? • How? What reassurances, beyond* what you've heard already, would be needed on: ○ How data is governed/ managed ○ How data is stored and accessed **Q11: How would you expect this to be communicated?**		Facilitator to note key points on the flip chart divided into expectant & new parents on one side/ society on the other Highlighting any priority points.	Getting into the detail of transparent research processes and communications what's good/ what's challenging and what they expect/ hope

Workshop 2, round 2 – in person					
Time	Agenda	Process	Who?	Process Tools	Expected Outcomes
(15 mins) 3:40-3:45 (5 mins)		To new and expectant parents? To wider society? Who should be communicating about this? Summary flip charts created (if time) * Trusted Research Environment access/ monitoring of use.		Flip chart: Summary points: 1, 2, 3	
3:45-4:00 **(15 mins)**	Menti.com Wrap up and close	Back to working as a whole group QM5: One thing that you have learnt or has particularly interested you from today's discussions. QM6: Advice you want to give to Genomics England as they develop research access to the Newborn Genome Programme. Reflections from Observers/ team on the day. Share the evaluation form. Your thank you payments will be settled within the next two weeks when we have your bank details. Thanks for being with us today and for all your hard work with us.		Menti	Summing up and clarity on next steps.
4pm	**Close**				