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# A public dialogue on genomic medicine: time for a new social contract?

Final report

Ipsos MORI



UK Research  
and Innovation

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## Foreword

We are delighted to see the publication of this important and timely report. It is increasingly clear that developments in genomics and other emerging forms of data-driven medicine have the potential to significantly improve human health and to inform more effective and efficient health systems. At the heart of these developments is a recognition that the ability to provide the best medical care for individual patients can be greatly enhanced by comparing their data with that of many others. That is, through the faster sharing of knowledge. It is also clear that a modern and effective health service capable of offering best care will be one in which clinical practice and data-driven research are highly integrated.

These benefits, which depend upon the collection, storage, and sharing of patient information are only fully achievable and sustainable in the context of well-founded public trust and confidence. That is, where patients, the wider public, health professionals, and researchers have shared expectations – a ‘social contract’ – about what constitutes reasonable and acceptable uses of patient data and samples. Any sustainable ‘contract’ of this kind will need to take the form of a relatively stable but nonetheless living dialogue capable of responding to the changes in medical science and technology and our shifting public attitudes and values.

This report presents the results of an inclusive and thorough process of public dialogue and makes a vital and timely contribution to ongoing discussions in this area and to the achievement of the conditions for the successful and sustainable development of the NHS Genomic Medicine Service. It explores the perspectives of members of the public, patients, and experts from a range of public, private, and charity organisations to the collection, storage and use of health data for genomic medicine. It reveals that the relationship between the National Health Service, patients, and the public is currently understood in terms of three core values: reciprocity, altruism, and solidarity. This is welcome evidence of the endurance and continued relevance of values informing the original establishment of the NHS as a research active organisation. The findings of this dialogue suggest that these values are likely to continue to inform understanding of the appropriate relationship between medicine, research and society as genomic medicine plays a more central role. This report also emphasises the importance placed by participants on equity in both access to and outcomes of health services as a key component of the social contract for genomic medicine.

There is no doubt that one of the most important factors in the recent success of the 100,000 Genomes Project has been the embedding of ethics support and advice, and patient/participant involvement throughout the organisation and at all stages of the project. The Participant Panel and Ethics Advisory Group have played crucial roles in establishing and maintaining public confidence in the project. The findings published in this report help to illustrate why this might have been the case.

The report is essential reading for everyone with an interest in the successful and appropriate establishment and future development of the NHS Genomic Medicine Service and of genomic and data-driven medicine more broadly. Given the profound importance of these developments and of



**Professor Dame Sally C Davies**

Chief Medical Officer for England and Chief Medical Advisor to the UK Government



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the findings of this report, we very much agree with the authors' recommendation that the ways in which genomics adds to and changes the social contract be embedded into the NHS Constitution

# Acknowledgments

This public dialogue on genomic medicine was co-funded by Genomics England, the Sciencewise programme<sup>1</sup>, and by the Scottish Genomes Partnership<sup>2</sup>. The authors would like to thank Genomics England, the Scottish Genomes Partnership and Sciencewise for their support and advice, and the members of the Oversight Group (OG), which was supported by Genomics England, for all their invaluable contributions throughout the study.

We would also like to thank all the public participants, clinicians, researchers, genomics counsellors, stakeholders and OG members who contributed to and reviewed materials, attended the dialogue events and were willing to discuss their hopes and priorities for genomic medicine in the NHS.

We would also like to thank Anna MacGillivray, the project evaluator, for her constructive formative feedback through the course of the study.

Our utmost thanks go to the 100,000 Genomes Project Participant Panel for their time and attendance at the dialogue workshops.

**Table 1: Members of the Oversight Group**

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11	Nick Hillier	Director of Communications	Academy of Medical Sciences

<sup>1</sup> Sciencewise is funded by UK Research and Innovation (UKRI). The Sciencewise programme aims to improve policy making involving science and technology across Government by increasing the effectiveness with which public dialogue is used and encouraging its wider use where appropriate to ensure public views are considered as part of the evidence base. It provides a wide range of information, advice, guidance and support services aimed at policy makers and all the different stakeholders involved in science and technology policy making, including the public. Sciencewise also provides co-funding to Government departments and agencies to develop and commission public dialogue activities.

<sup>2</sup> The Scottish Genomes Partnership is funded by the Chief Scientist Office of the Scottish Government Health Directorates [SGP/1] and The Medical Research Council Whole Genome Sequencing for Health and Wealth Initiative (MC/PC/15080).

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8	Dr Geraldine Clarke	Senior Data Analyst	The Health Foundation
9	Dr Sophia Skyers	Director, CIBS IQ Research	CIBS IQ Research
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15	Monika Preuss	Head of Genomic Science, Genomics and Emerging Technologies	Department of Health and Social Care
16	Pauline Mullin	Head of Communications	Medical Research Council
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# Executive summary

## 1. Introduction: a dialogue to capture public views of genomics

The NHS Constitution reminds us that our health service is founded on a common set of principles and values that bind together patients, the public and staff so that it is effective and equitable. It recognises that each party has roles and responsibilities and therefore the Constitution can be seen as a form of **'social contract'** which aims to bring the highest levels of human knowledge and skill to save lives and improve health.

The Chief Medical Officer for England's 2016 Annual Report, **'Generation Genome'** argued for a re-thinking of the wider **social contract** in healthcare today, taking into account four areas<sup>3</sup> of medical and research practice. It also provided a public statement on the purposes and benefits of genomic data in the NHS. Chapter 16<sup>4</sup> of the report draws together the important requirements that need to be in place if genomic medicine is to be ethically and socially acceptable to the UK population.

The purpose of this dialogue was to take into account members of the public's priorities and concerns when considering **how genomics might alter or affect the social contract in healthcare**. This was in order to inform the way genomics should be mainstreamed in the NHS in England, as well as to inform healthcare policymaking in Scotland, and the future of genomics more broadly.

### Why was a Sciencewise public dialogue the most suitable approach to canvass the public's views on a genomics future?

A Sciencewise public dialogue provides in-depth insight into citizens' views, concerns and aspirations on issues relating to science and technology<sup>5</sup>. These issues are often complex and unfamiliar to citizens and therefore their exploration is better suited to a qualitative approach.

### How are this dialogue's findings valid?

Applying criteria used in the social science literature<sup>6</sup> to determine the credibility of qualitative research findings, we can be confident that the principles and views presented here are credible and valid due to the following strategies used in this dialogue:

- a) **accounting for researcher bias** – in order to ensure the information given to the dialogue participants was balanced, we engaged with a diverse range of expert perspectives, including data privacy campaign groups, all of which fed into the development of the research materials and stimulus.
- b) **accounting for sampling bias** – the dialogue participants were recruited to be reflective of the wider UK population, using quotas informed by UK census data.
- c) **accounting for research bias** – participants were given all the information to enable them to develop their views, and given time to reflect between events. Experts rotated to each discussion group to ensure the participants were exposed to the same sort of information. Plenary sessions were built in to each event so we were able to identify a range of views including commonalities and outliers.

<sup>3</sup> The four areas are: consent, confidentiality and the availability of the best care for patients and families, obligations of health professionals, lab staff and researchers; system responsibilities.

<sup>4</sup> Lucassen, A. Montgomery, J. Parker, M. (2016) 'Ethics and the social contract for genomics in the NHS' in Annual Report of the Chief Medical Officer 2016: Generation Genome (Published by the Department for Health and Social Care).

<sup>5</sup> <https://sciencewise.org.uk/about-dialogue/what-is-public-dialogue/>

<sup>6</sup> <https://ebn.bmj.com/content/18/2/34>



- d) **meticulous record keeping and systematic analysis** – a transcript was produced from each discussion group, with each one analysed using qualitative social science technique. A series of analysis sessions attended by the Ipsos MORI team, the independent evaluator and Genomics England has ensured that all the different perspectives are represented in this report.
- e) **participant and expert validation** – bringing participants back to a final genomics summit enabled us to explore key issues in further detail and thereby validate the views we noted in previous workshops. Experts who observed and participated in the dialogue discussions have reviewed the report and given feedback.
- f) **data triangulation** – the views and perspectives of the dialogue participants are supported by the rapid evidence review, and other relevant research, all of which are referenced throughout this report, where appropriate.

**Are these findings representative / generalisable?** This dialogue sought to understand the depth and complexity of views on a genomic medicine service and the impact of information on these views. In contrast with quantitative approaches, dialogue findings are not statistically significant. However, dialogue enables us to explore the values and reasons underlying the views expressed.

**How is this public dialogue useful to decision-makers?** A dialogue is a valid and robust way to inform policy, especially to inform understanding of the range of options open to policy makers which will fit with the sensibilities of the public; and why the public think the way they do. The culmination of this public dialogue is this report which provides detailed and nuanced evidence on how citizens' views, concerns and aspirations can be operationalised in a genomics future.

Ninety-seven members of the public, and thirty experts came to evening and reconvened day-long Saturday events in Coventry, Edinburgh, Leeds, and London. A proportion of each group was reconvened to a final Genomics summit event in London (n=23 in total), where the group was again joined by experts. A total of forty-three experts attended the dialogue workshops and the Genomics summit. The sessions were facilitated by a team from the Ipsos MORI Public Dialogue Centre.

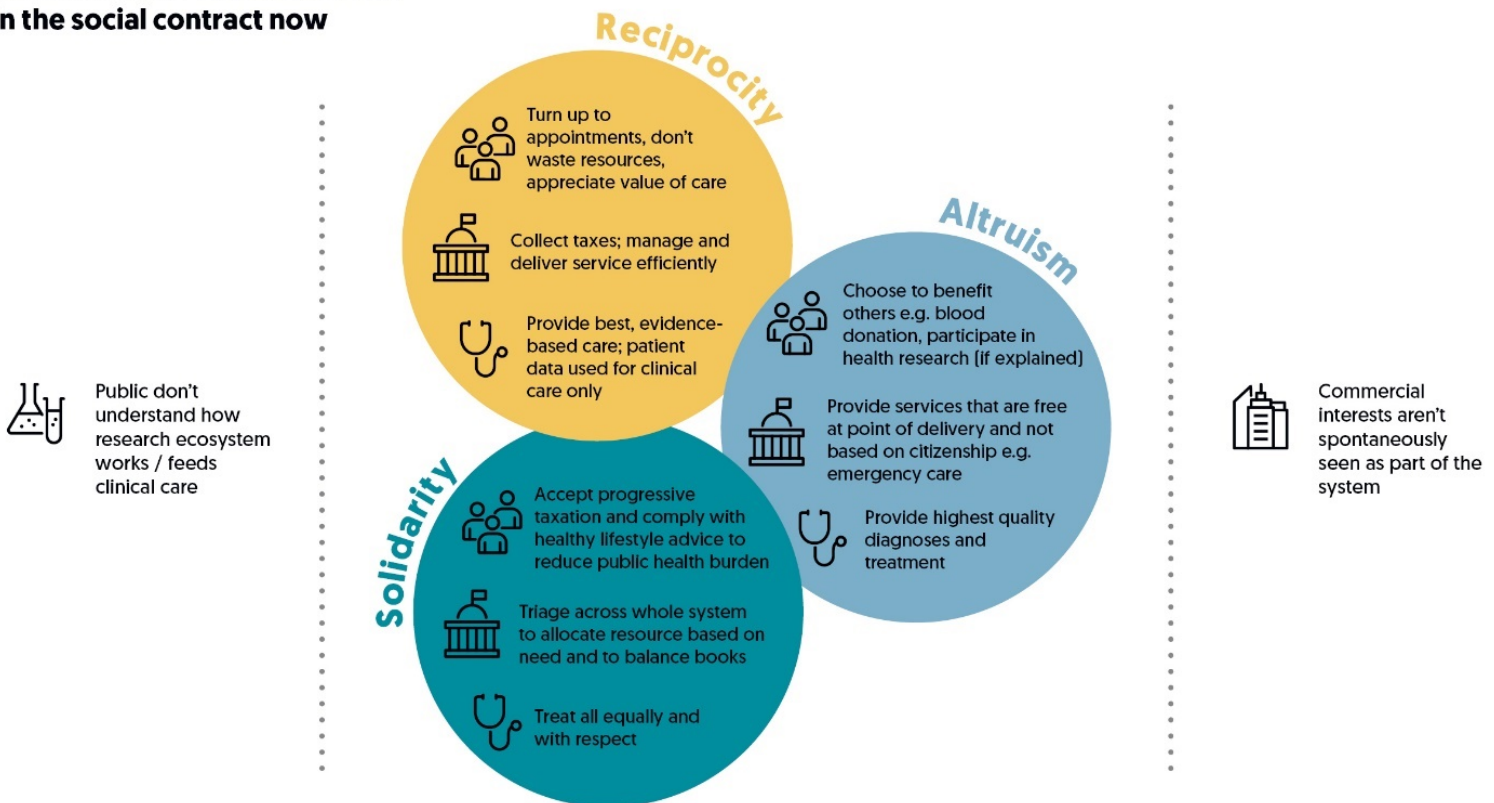
A **rapid literature review** was conducted to inform the dialogue materials and to ensure that the project built on the work of previous social research on attitudes to genomics. The structure of the dialogue was developed in partnership with the Oversight Group, and materials and key questions were workshopped prior to the dialogue workshops with an external group of 15 stakeholders reflecting a range of perspectives.

## 2. Public views of the healthcare social contract

The dialogue participants were not familiar with the term 'social contract' or the explicit concept to which it refers. However, when participants discussed during the dialogue how they think healthcare works now, and how genomics should operate in society, their responses were predicated on assumptions about the **social contract** that they believe to be in place. We are therefore able to draw out the ingredients they feel this contract should have. Chapter 2 discusses this in detail.

First, they had some clear perceptions of how they felt the social contract works today (before considering the idea of genomics). They saw it including three elements; **reciprocity, altruism, and solidarity**.

## Public views of key behaviours in the social contract now



**Reciprocity** meant a 'transactional relationship' in which users "give" and "get". Expectations were:

- The NHS will provide **evidence-based care**.
- Patients are given **diagnoses and information about their condition**, and the chance to discuss this with their **doctor**.
- Doctors are trusted to **only share patient information and data with those who are directly involved in a patient's care**.
- The Government's role is to **manage and deliver healthcare services efficiently**, through taxes collected centrally.
- Users (patients and the wider public) are expected to **behave well** (e.g. to not abuse NHS staff), to **value resources** (e.g. turn up for appointments on time) and **support the NHS**; understand and appreciate the value of the care given.

The social contract also requires **altruism**:

- Members of the public are expected to **want to benefit others as well as themselves**, and behave altruistically (e.g. donating blood, letting ambulances through traffic).
- Participation in health research (when understood) was seen as an altruistic act, as it often does not deliver immediate individual benefits.
- The NHS should provide healthcare services that are **free at the point of delivery**; regardless of a person having UK citizenship; there was **an inbuilt moral stance that it would be wrong to deny care to anyone**, especially emergency care.

**Solidarity** was the third key aspect of the social contract, requiring:

- A shared acceptance of paying for healthcare by **progressive taxation**.
- Public acceptance that individual good health contributes to the public good, and complying with health instructions (e.g. vaccinations) to reduce the public health burden.
- The NHS is expected to carry out **effective triage**<sup>7</sup>, so that patients priority for treatment is based on clinical need.
- Clinicians exercising a **moral and ethical duty to treat everyone equally and with respect**.

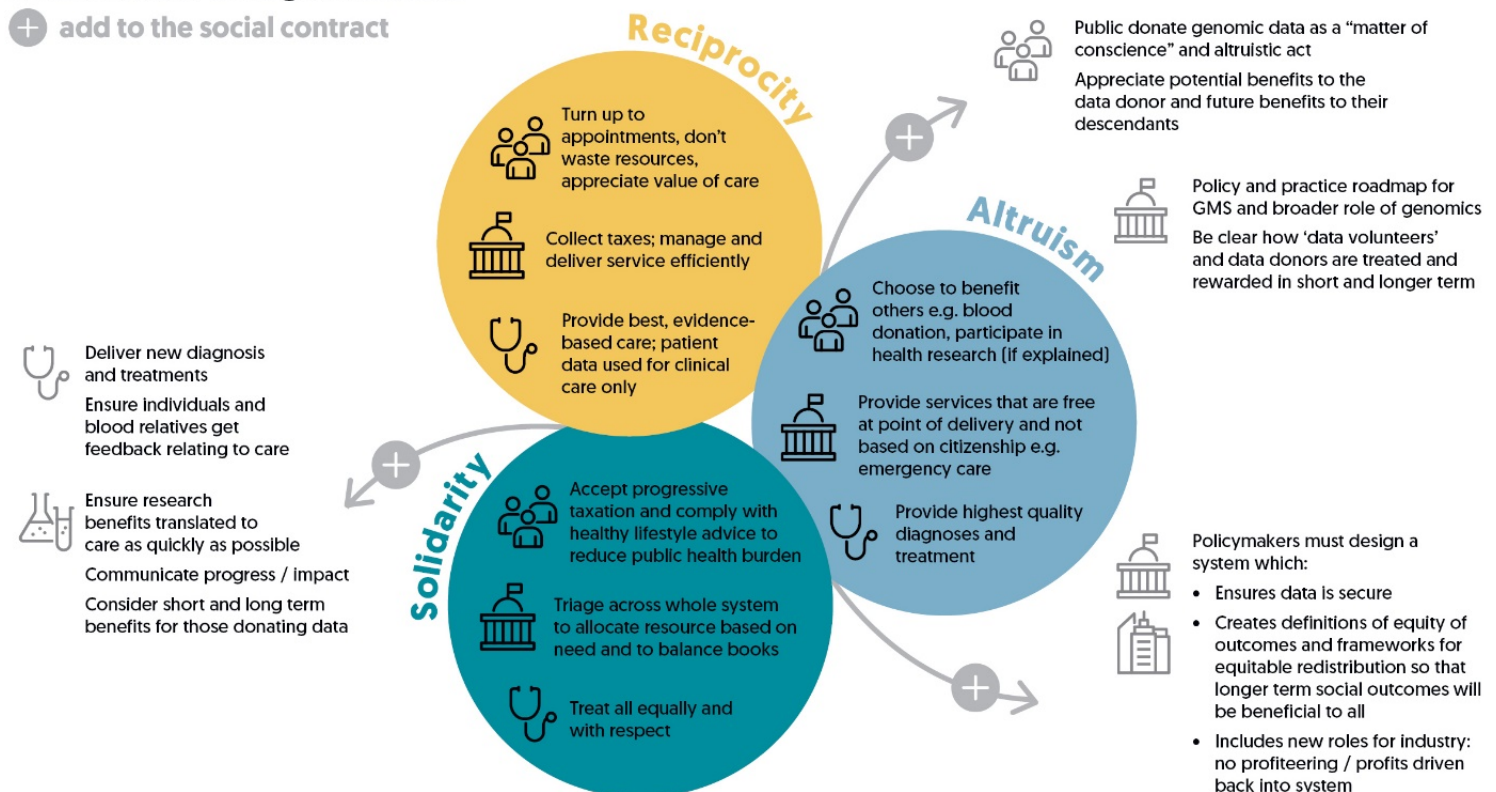
Importantly, in dialogue participants' conception of the social contract, **expectations of researchers were often absent; and the role of commercial companies was ignored**. This reflects the fact that the general public are generally not aware of the role biomedical research plays in the healthcare system, or the role that charities, industry, and others play in the existing system.

### 3. What genomics might add to the social contract

By the end of the dialogue, participants had discussed the implications of genomics for the social contract. Overall, in a genomics-driven healthcare of the future, they felt the principles of reciprocity, altruism and solidarity would need to remain core to the social contract, and require largely the same behaviours of all the actors.

However, the new science of genomics means that the **three elements might intersect in new ways** - leading to **new expectations of behaviours of all actors in the system**.

#### Public views of what genomics will



<sup>7</sup> The category assigned to a patient as a result of an initial assessment by medical or nursing staff in an Accident and Emergency Department. The **triage** category is used to determine the patient's priority for treatment, and to inform the patient of their waiting time.

- **Reciprocity ← → Altruism:** Analysing an individual's genome requires that data has been donated by many. This is because an individual's data needs to be compared against that of others to properly inform their diagnosis and treatment. This blurs the line between reciprocity and altruism. The public in the dialogue saw giving their data as an altruistic act - but also expected benefits would probably accrue to their families and descendants.
  - There would, therefore, be an increased expectation that the public would donate data, but participants felt that data donation should not be mandatory.
  - Participants thought that government and healthcare policy makers would need to create a roadmap for genomics where the status of data donors is made clear, and any rewards for them carefully worked out.
- **Reciprocity ← → Solidarity:** Genomics can use patient data in research and clinical care at the same time, creating a feedback loop between both to increase possible learning. Participants felt that this **research/clinical blurring** brought the ideas of reciprocity and solidarity closer together.
  - Both clinicians and researchers in a genomics future would be responsible for ensuring that patients and families are informed about the progress of research.
  - They should also focus on translating the benefits of research into care, to help all of society, as quickly as possible.
  - Both researchers and clinicians should communicate to the public what they are doing with genomic data, and the link between their work and making discoveries with clinical impact; so the public can know their collective action is worthwhile and having impact.
- **Solidarity ← → Altruism:** In the future, if large genomic datasets exist, and can potentially be accessed by new actors (such as commercial companies) social solidarity will be affected by the altruistic acts of individual donors. Dialogue participants felt that:
  - Policy makers should take the long view – design a future which prevents dystopian social outcomes. Participants emphatically did not want their donation of genomic data used outside of healthcare and research in ways that would create a stratified society which disenfranchises vulnerable members. (e.g. using predictive genomic testing when a person applies for insurance) or different racial or ethnic groups.
  - There would be a new role for industry in bringing the benefits of genomics to society; but either voluntary or external regulation will be needed. Again, participants did not want to see the altruistic data donations of individuals used to enable private sector profiteering.

#### 4. What genomics might change about the social contract

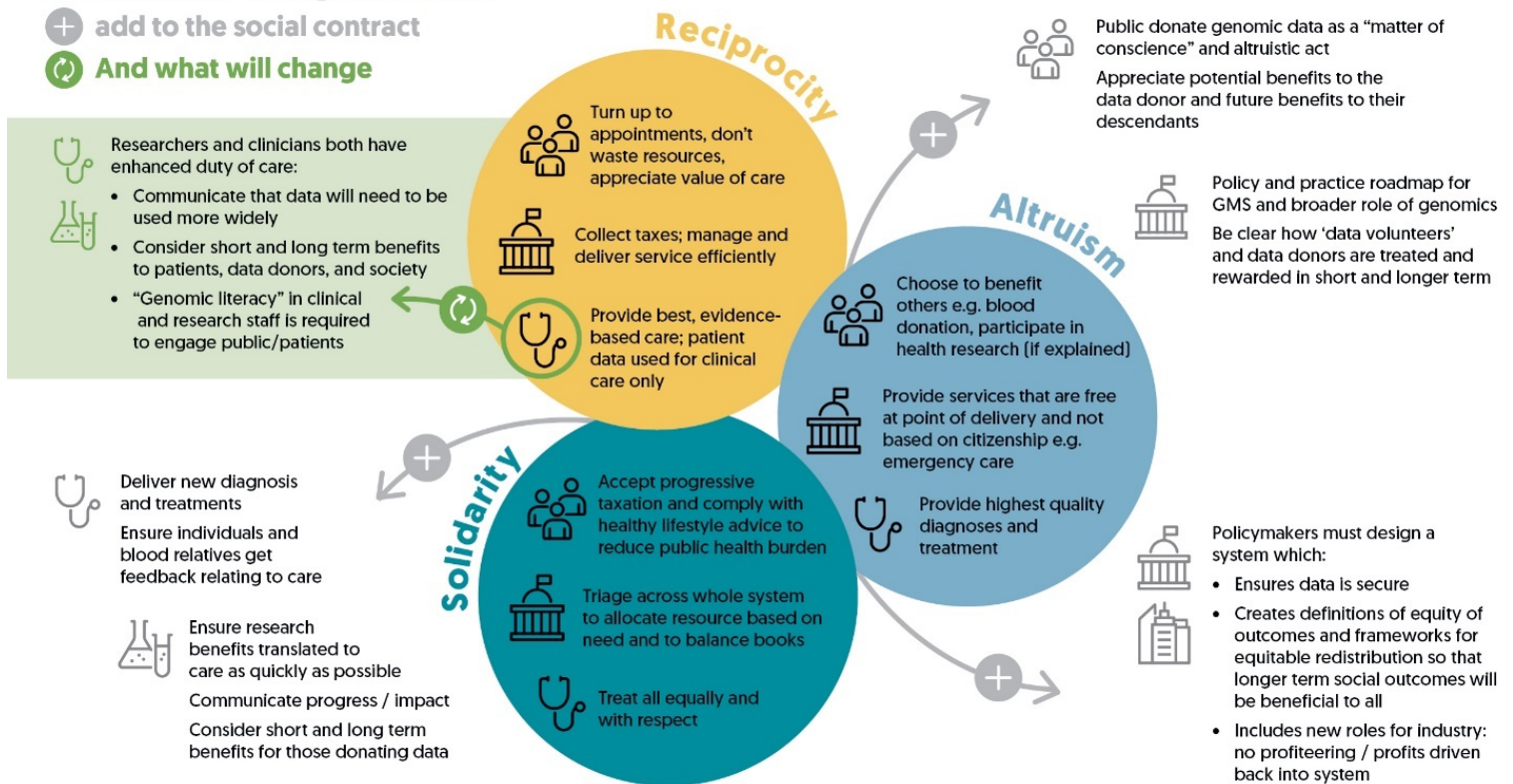
As well as these additions, participants also felt there would be one change to the social contract; **genomics changes the expectation that clinicians will only share data with those involved directly in patient care.** This leads to new duties of care for both researchers and clinicians.



## Public views of what genomics will

+ add to the social contract

And what will change



- Participants wanted to empower **clinicians to make the final decisions on disclosing information** (for instance, to family members affected by test results). However, participants acknowledged that this was not an easy recommendation to make, and often changed their views on whether there should be a general policy or whether clinicians should judge on a case-by-case basis. On one hand they wanted decision-making to be shared with patients and very transparent, and they wanted patients to be able to challenge decisions they did not agree with. Yet on the other hand, they did not want to add any administrative burden on clinicians from a medico-legal perspective, which making such decisions would involve.
- After deliberation, participants were broadly happy to accept the **uncertainty** of not knowing what information genomics might reveal, and **trade off complete confidentiality** against potential benefits for themselves and others, as part of their broad consent to participate in genomics research.

Participants felt both clinicians and researchers should be equipped with "**genomic literacy**" to support the informational, emotional and practical needs of patients and data donors; giving more information and support than has been necessary in the past when communicating with research participants.

## 5. Red lines: unacceptable uses of genomic data

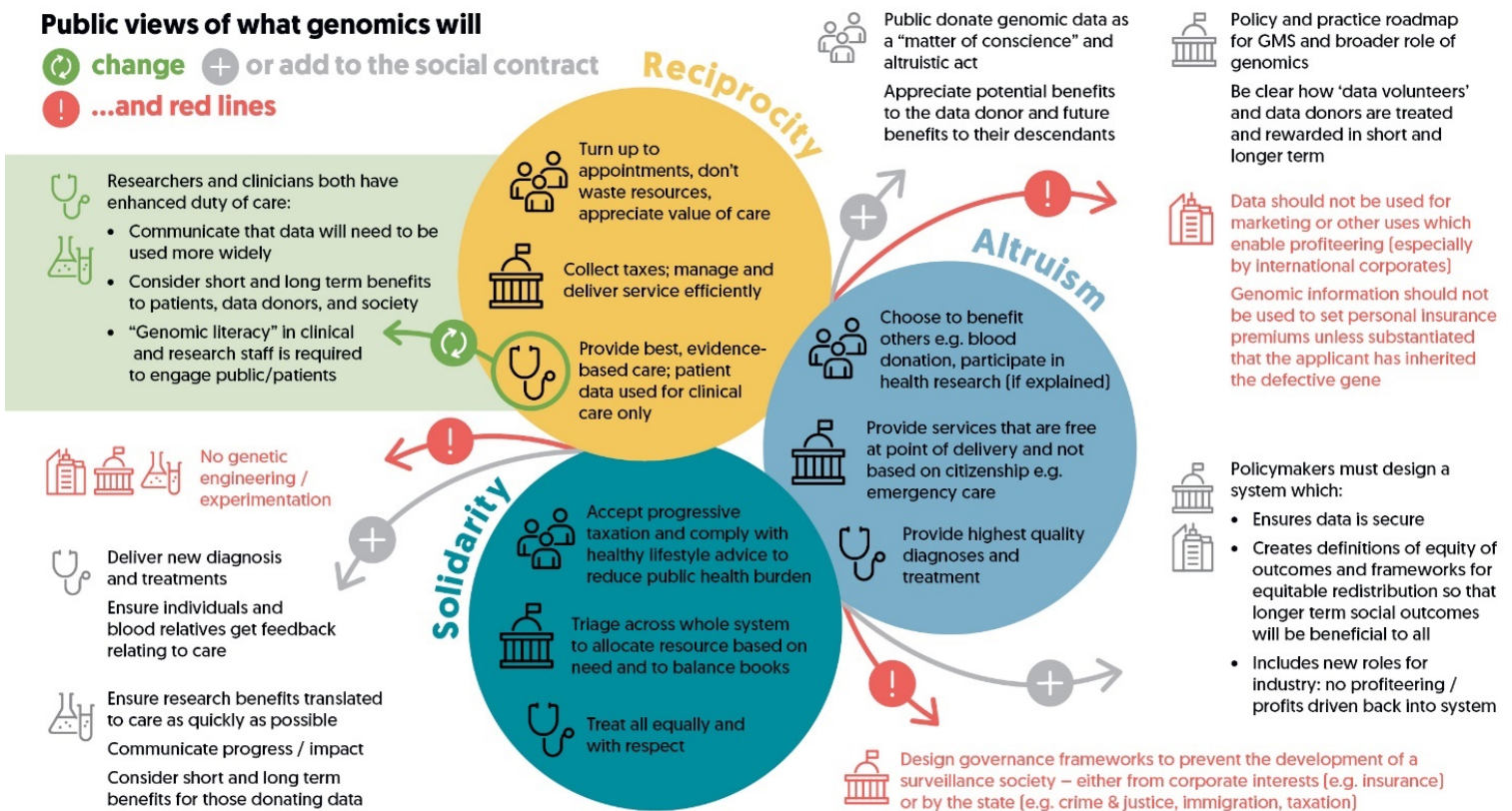
Participants had some clear limits for how far they thought genomic data, and information derived from genomic analysis, should be used.

- **Genetic engineering:** Participants saw huge ethical issues around editing the human genome, especially to enhance human capabilities. They wanted to prevent this happening, at least without much more public engagement around the uses of genomic data.
- **Surveillance society:** many participants had a sense of fatalism that the day would come when genomic data would be used, either by corporate interests (e.g. insurance) or by the state (e.g. crime and justice and taxation) to stratify society in order to identify and penalise individuals with acute healthcare needs, and unfairly monitor groups in society; they wanted to prevent this.
- **Administrative and political uses:** there was a general feeling that more data is being collected all the time which has the potential to be used in ways which reduce access to state funded services such as welfare, and create a general sense of control in society; participants wanted to prevent this.
- **Predictive insurance tests:** participants were not supportive at all of using genomic information to set personal insurance premiums
- **Targeted marketing:** participants were very clear that data should not be used for marketing, or other areas of product development which might enable profiteering, especially by international corporates.

Participants wanted assurances from policy makers, the government, as well as independent stakeholders (e.g. data privacy campaign groups) that there is a robust governance framework in place which ensures their red lines don't happen in practice, which incorporates a consent process that makes it clear what researchers intend to do with genomic data.

**Public views of what genomics will**

**change + or add to the social contract**  
**! ...and red lines**



## 6. Communicating genomics as a collective endeavour in healthcare

This dialogue suggests that the public are likely to be open and welcome to the idea of using genomics more widely in healthcare.

- Initially, participants had a **limited understanding of genomics**; almost nobody had heard of a 'genome' or 'genomics'. When introduced to the ideas, however, almost all responded positively and many developed high **expectations of genomics**, envisaging a near-term future with new treatments and personalisation of care, and significant cost savings for the NHS. Others were initially worried about who would be using their genomic data and for what purposes even though they could recognise the benefits of their genomic data being used in health research.
- **Almost all were relaxed about their health and genomic data being used in health research.** This support is, however, conditional on consent being obtained first; the use of de-identified data only and red lines being respected; robust risk assessment and safeguards being implemented and maintained by policy makers, researchers and clinicians; and genomics seen to be having real clinical impact.

Realising the potential of genomics in the longer term will require a **critical mass of UK citizens supporting it to the extent they are willing to participate**. While this is a longer-term aim, creating and implementing a genomics narrative should be an immediate priority as the dialogue participants were clear that policy makers and the NHS have a responsibility to do more to inform people about genomics.

The narrative on genomics will need careful framing, because, if the benefits of genomics are over-hyped, or the uncertainties or limitations downplayed, there is a **risk of fracturing the public's trust in genomics**, and in health research more generally. Communications should convey the idea that genomics is an **ambitious, potentially ground-breaking programme of activities** that aims to:

- redefine what best care is: for example genomics is moving away from "one size fits all" treatments to patient care tailored to their unique genetic make-up
- deliver new medical discoveries
- but also change healthcare in ways which cannot be predicted with certainty.

**It should not be** confused with a service like blood or organ donation; a key misunderstanding in discussions. Instead, communicating the idea of a "**national moonshot**" or shared endeavour may work to link genomics to a wider UK programme of innovation in which we are all involved.

**In the longer-term** as genomic medicine is scaled-up, genomics will need a call to action - if a social contract around genomics is going to work the public need to know what genomics is and be motivated to take part, so there is a need for a mission statement that conveys the idea that success needs solidarity, altruism and reciprocity. We can infer from the participants' discussions that the following statement might work as a call to action, as well as communicate the essence of the social contract in an intelligible way.

**"People powered genomics".**

As genomics evolves, further public and stakeholder engagement may be needed to ensure the social contract remains relevant and useful.



# 1 Introduction

Genomics England with support from Sciencewise, commissioned Ipsos MORI to undertake a series of public dialogue workshops exploring how the public feel that genomic medicine should best be 'mainstreamed' into the NHS. The project involved exploring the principles the public believe should underlie any 'social contract'<sup>8</sup> between the public and providers of genomic medicine.

## 1.1 What is genomics?

Over the last decade, genomics has rapidly accelerated as an area of clinical relevance, with major advancements within our diagnostic capabilities of rare diseases, alongside greater efficacy when prescribing medication. The future potential of genomics is huge, with increasingly accurate and sophisticated diagnostic techniques alongside better therapeutic possibilities for many diseases.

## 1.2 Genomics in the NHS and the Generation Genome report

In 2012, the Department of Health and Social Care launched the 100,000 Genomes Project and founded Genomics England<sup>9</sup>, as an independent company whose sole shareholder is the Secretary of State for Health, to coordinate the project. The aim of the 100,000 Genomes Project was to collect and analyse 100,000 genomes from 70,000 people. Those invited to take part were patients with certain kinds of cancers, and rare<sup>10</sup> or unknown diseases. In those with cancers, a genomic sample was taken from both healthy (non-cancerous) cells and cancerous cells. The Project reached its target of sequencing 100,000 genomes in December 2018.<sup>11</sup>

As a result of the success of the 100,000 Genomes Project, NHS England is establishing infrastructure to **mainstream genomic medicine across the NHS**. The genomic testing available and the criteria for access is defined as part of the National Genomic Test Directory<sup>12</sup>. **Genomic tests are also available to patients in Scotland, Wales and Northern Ireland** through different NHS systems.

A key aspect of the infrastructure in England is seven Genomic Laboratory Hubs that will co-ordinate and deliver genomic testing for patients covering defined areas of the country. In addition, the UK government has recently announced its intention to analyse a further five million genomes, as part of its effort to grow the database upon which genomic research can draw.<sup>13</sup>

The Chief Medical Officer for England's 2016 Annual Report, '**Generation Genome**', sought to explore the potential of genomics within the health and social care system. The report provides a public statement on the purposes and benefits of genomic data in the NHS. It argues for a re-thinking of the wider social contract in healthcare today, taking into account four areas<sup>14</sup> of medical and research practice.

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<sup>8</sup> To ensure the dialogue participants could engage with the concept of a social contract for genomics, a working definition was formed for the purpose of this dialogue: *"The shared, reasonable assumptions and expectations that everyone in the system has, which guide how the [genomics] system works"*

<sup>9</sup> <https://www.genomicsengland.co.uk/>

<sup>10</sup> For patients with rare diseases, their parents also had their whole genome sequenced.

<sup>11</sup> <https://www.genomicsengland.co.uk/the-uk-has-sequenced-100000-whole-genomes-in-the-nhs/>

<sup>12</sup> <https://www.england.nhs.uk/publication/national-genomic-test-directories/>

<sup>13</sup> <https://www.gov.uk/government/news/matt-hancock-announces-ambition-to-map-5-million-genomes>

<sup>14</sup> The four areas are: consent, confidentiality and the availability of the best care for patients and families, obligations of health professionals, lab staff and researchers; system responsibilities.

In the light of this report, Genomics England with support from Sciencewise considered it important to canvass the views of the public on their hopes and priorities for genomics, as well as their views on the potential changes to, or evolution of, the social contract.

### 1.3 The dialogue aim and objectives

The overall aim of this dialogue was to engage the public in a dialogue to gain a greater understanding of the public aspirations, concerns and expectations of genomics and genomic medicine.

In this context, the **public dialogue sought to explore the following objectives.**

1. Establish current knowledge and understanding on public views and attitudes to i) genomic medicine, ii) whole genome sequencing iii) the concept of a 'social contract' between the public and the NHS and iv) safeguards/'red lines' essential to public support/trust for genomic medicine, through a review of previous dialogue, engagement, consultation and related research projects involving the public.
2. Understand participants' aspirations and concerns around the use of genomic data and other personal information.
3. Understand how participants 'trade off' concerns about data, privacy and use of their information against potential health and other benefits that may come from genomic medicine, and what safeguards need to be in place.
4. Explore understanding of the idea of a 'social contract' as applied to the NHS and the principles by which it is understood to, or should, operate.
5. Explore what expectations and understanding are shared between patients, the public, clinicians, academics, industry and other stakeholder groups around the ambitions and outcomes for genomics and genomic medicine in the UK.
6. Contribute to and inform the ongoing policy, political and ethical environment for genomic medicine to flourish, as the benefits and opportunities presented by the technology are realised.
7. Contribute to the wider policy, political and ethical environment about the collection and use of data by government beyond healthcare and genomics.
8. Develop an understanding of the language and terms that the public and other stakeholders use in association with genomic medicine in order to inform communications for and about genomic medicine activities and services in the UK.

### 1.4 Study design

The dialogue was intended to build on existing research on public attitudes to genomics, and fill gaps in existing understanding. As such, Ipsos MORI conducted a **rapid literature review**<sup>15</sup> in August 2018, which showed what is already known by the public, and also the views of experts, clinicians, and

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<sup>15</sup> <https://www.genomicsengland.co.uk/what-do-we-know-about-public-attitudes-to-genomics/>

patients on some of the ‘live issues’ in genomics. The findings were shared with the Oversight Group and Stakeholder Group (see below) and feedback was used to inform the materials used in the dialogue workshops.

An **Oversight Group (OG)**<sup>16</sup> provided guidance to the dialogue: the group was comprised of clinicians, academics, policy makers, patients and representatives from a range of public, private and third sector organisations who play a part in genomics. The group’s role was to help shape and steer the project, and use their collective expertise to advise on the technical, ethical and practical issues associated with genomics.

In September 2018, Ipsos MORI organised a **Stakeholder Group**<sup>17</sup>. This group contained a broader range of experts and stakeholders within genomics and helped capture a greater diversity of voices within the area. Their views, along with those from the Oversight Group, helped ensure the information presented to the public was balanced and technically accurate.

**Ninety-seven members of the public, and thirty experts** came to evening and reconvened day-long Saturday events in Coventry, Edinburgh, Leeds, and London. These locations were chosen to reflect the diversity of geographical areas in England and Scotland. The workshops took place between October and November 2018.

**Participants were recruited to be reflective of the population of each of the four areas.** To achieve this, we designed a quota for gender, age, ethnicity, working status, and socioeconomic grade. Note that this sample should not be taken as *representative* from a statistical point of view. Instead the mix of participants aimed to *reflect* the mix of people in and around each location.

Public participants were given a cash honorarium. This is standard in Sciencewise dialogues and is done to ensure that a diversity of participants are able to attend the event regardless of financial circumstance. For this reason also, the day-long events were held on a Saturday.

At each location we met with the participants twice: at an evening event where we introduced genomics, and a full-day long workshop where we could explore the material in greater depth. Each table at the workshops was facilitated by the team at Ipsos MORI.

The **workshops in Edinburgh** took place in a slightly different context. The key difference between Scotland and England is that the NHS Scotland and its partners<sup>18</sup> are considering how to make best use of genomic testing in the future. Genetic tests already take place routinely in NHS Scotland, including exome testing<sup>19</sup>, but the efficacy of whole genome testing is still being assessed through research. Canvassing public views is an important part of this work.

#### 1.4.1 The structure of the workshops

Informing the dialogue participants about genomics, genomic medicine, and the different actors who will play a part in it was an important first step, given the limited understanding of genomics among the UK population (see Section 3.1).

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<sup>16</sup> A full list of Oversight Group members is included in the Acknowledgements section.

<sup>17</sup> This included patient group representatives, data privacy campaign groups, social scientists, and representatives from pharma and med-tech companies.

<sup>18</sup> Such as Scottish Government and Scottish Genomes Partnership.

<sup>19</sup> Next-generation sequencing techniques “read” all of the DNA in a person’s genes letter by letter (all 3 billion of them) to allow comparisons to be made with the DNA of other people. Exome sequencing is the targeted analysis of the gene regions that code for protein. Whole genome sequencing examines the entire genetic code of an individual. As researchers learn more about what individual DNA changes can mean for health and disease, information provided by these techniques will be used by the NHS to provide new diagnoses and treatments.

Day-long follow-up sessions exposed the dialogue participants to a range of expert perspectives with regard to the benefits which may arise from genomics, its uncertainties, and potential risks, as well as the 'live' ethical and practical issues such as consent and patient confidentiality,

Engaging the dialogue participants through extended and staggered events meant they were able to deliberate and develop more considered opinions on the usage of genomic data, the responsibilities and obligations of different genomic actors, and the assurances/messages which are likely to encourage genomic data donation.

At each location interactions with participants consisted of the following:

<u>Evening event</u>	<u>Day-long workshop event</u>
<ul style="list-style-type: none"> <li>• Discussion around healthcare – hopes and priorities</li> <li>• Plenary presentations and table discussions to introduce genomics and genomic medicine</li> <li>• Discussion of hopes and fears on genomics / genomic medicine</li> </ul>	<ul style="list-style-type: none"> <li>• Genomics expert presentations</li> <li>• Scenario-based family trees (using visual illustrations). These different family trees allowed the public to engage with abstract ethical and practical issues associated with genomics</li> <li>• Discussion of a social contract in genomics</li> <li>• Discussion of genomic data: consent, confidentiality, security and safeguards, and red lines</li> </ul>

#### 1.4.2 The role of experts in this dialogue

Alongside members of the public, a number of **experts were invited to each event**. They were all people with significant experience of genetics/genomics either as clinicians, researchers, or genetic counsellors. Their role was to help clarify any technical questions asked by dialogue participants, explain what practical and ethical issues exist in genomics, how those issues are handled, and engage alongside the public in discussions about a new social contract for genomics. In addition it was an opportunity for them to directly hear some of the issues raised by public participants.

Having **participants from the 100,000 Genomes Project** join in the dialogue workshops engaged the public with the lived experience of having a genetic condition, and the idea of familial implications. Their input brought a sense of realism to the high expectations that participants had for genomics at the outset.

**Table 3: Experts who attended and contributed at a dialogue event**

Name	Role	Organisation	Event location
Simon Wilde	Communications Manager	Genomics England	London event 1
Tom Fowler	Deputy Chief Scientist and Director of Public Health	Genomics England	London event 1
Sarah Caffery	100,000 Genomes Project Participant	N/A	London event 1
Dr. Lea-Rebecca Lahnstein	Genomics England Clinical Interpretation Partnership (GeCIP) Cross-Cutting Coordinator	Genomics England	London event 1
Christine Patch	Consultant Genetic Counsellor	Department of Clinical Genetics at Guy's & St	London event 2

		Thomas' NHS Foundation Trust	
Dr Richard Scott	Consultant in Clinical Genetics, Head of Rare Disease Programme	Genomics England	London event 2
Professor Tim Hubbard	Professor of Bioinformatics, Head of Genome Analysis	King's College London, Genomics England	London event 2
Lisa Dinh	Senior Communications Manager	Genomics England	London event 2
Simon Wilde	Communications Manager	Genomics England	Coventry event 1
Rebecca Middleton	100,000 Genomes Project Participant	N/A	Coventry event 1
Ann Dalton	Consultant Clinical Director	Sheffield Children's NHS Foundation Trust	Coventry event 1 and event 2
Dr Felicity Boardman	Assistant Professor	Division of Health Sciences, University of Warwick	Coventry event 2
Sean James	Arden Tissue Bank Manager & South Midlands Genomics Ambassador	University Hospitals Coventry & Warwickshire	Coventry event 2
Katherine Mazur	Genetic Counsellor	Birmingham Women's and Children's NHS Foundation Trust	Coventry event 2
Simon Wilde	Communications Manager	Genomics England	Leeds event 1
Dr Mushtaq Ahmed	Manager Genetic Counsellor	Leeds Teaching Hospitals NHS Trust	Leeds event 1
Kate Grafton	Principal Lecturer	Leeds Beckett University	Leeds event 1
Dave McCormick	100,000 Genomes Project Participant	N/A	Leeds event 1 and event 2
Andy Hart	100,000 Genomes Project Participant	N/A	Leeds event 1 and event 2
Prof Eamonn Sheridan	Professor of Clinical Genetics	University of Leeds	Leeds event 1 and event 2
Saghira Malik Sharif	Principal Genetic Counsellor	Leeds Teaching Hospitals NHS Trust	Leeds event 1 and event 2
Prof Colin Johnson	Professor of Medical and Molecular Genetics	University of Leeds	Leeds event 2
Dr Jon Fistein	Associate Professor in Clinical Informatics	Leeds Institute of Health Sciences	Leeds event 2
Cheryl L Stopford	Genetic counsellor	NHS England	Leeds event 2
Stephanie Hart	Genetic counsellor	NHS England	Leeds event 2
Prof Zosia Miedzybrodzka	Professor of Medical Genetics	University of Aberdeen	Edinburgh event 1
Prof David Porteous	Chair of Human Molecular Genetics & Medicine	Centre for Genomic and Experimental Medicine, University of Edinburgh	Edinburgh event 1
Dr Wendy Inglis Humphrey	Project Manager, Scottish Genomes Partnership	Centre for Genomic and Experimental Medicine, University of Edinburgh	Edinburgh event 1 and 2
Simon Wilde	Communications Manager	Genomics England	Edinburgh event 2

Dr Victoria Svinti MacLeod	Bioinformatician	MRC Human Genetics Unit, University of Edinburgh	Edinburgh event 2
Michelle McGachie	Bioethicist, PHD Scholar	University of Glasgow	Edinburgh event 2
Nicola Coates-Dutton	100,000 Genomes Project Participant	N/A	Edinburgh event 2
Joana Gomes	Genetic counsellor	NHS Grampian	Edinburgh event 2

### 1.4.3 A Genomics Summit

The Genomics summit took place in December 2018<sup>20</sup>. This event was an opportunity to create a lively, informed conversation between experts, patients and the public<sup>21</sup>, and explore areas in even greater depth than had been possible at the previous events. Participants rotated around five stations set up in a large room, each one exploring a particular aspect of genomics:

- Station #1: **Where is our data?** – participants were asked to reflect upon usage of genomic data, and what assurances are needed to encourage data donation.
- Station #2: **The patient of the future** – in light of the anticipated benefits arising from genomics as well as its uncertainties and limitations, participants deliberated on what behaviours they would expect from the different genomic actors.
- Station #3: **One big family** – given the genetic test result has familial implications, this station asked participants to consider the implications for consent and patient confidentiality, and how patient information and emotional needs should be met in genomic medicine.
- Station #4: **Show me the money** – this station explored what participants expect from genomic medicine both in terms of access and outcomes.
- Station #5: **Blurred lines** – this station looked at what people expect from clinicians and researchers (including those in commercial companies) given research and clinical practice come together in genomics

Participants were able to engage with a mix of stimulus materials (e.g. mocked-up newspaper headlines), real world resources (e.g. 100,000 Genomes Project consent forms); and props (e.g. a data 'black box'), before moving to the next station. After each section, final thoughts were written in a booklet prepared for the event.

**Table 4: Members of the Oversight Group and Stakeholder Group and experts who attended and contributed to the genomics summit.**

	Name	Role	Organisation
1	Vivienne Parry	Head of Engagement	Genomics England
2	Professor Jonathan Montgomery	Professor of Health Care Law	UCL Faculty of Laws, University College London
3	Dr Mark Bale	Deputy Head of Health Science and Bioethics	Department of Health
4	Dr Natalie Banner	Lead	Understanding Patient Data
5	Phil Booth	Coordinator	Med Confidential
6	Pete Mills	Assistant Director	Nuffield Council on Bioethics
7	Freddie Baker	Lead Policy Officer	Information Commissioners Office

<sup>20</sup> A total of forty-three experts attended the dialogue workshops and the Genomics summit.

<sup>21</sup> Participants (n=7 from each) from the London, Coventry and Leeds workshops were invited back to a half-day-long Summit which was held in London.

8	Monika Preuss	Head of Genomic Science, Genomics and Emerging Technologies	Department of Health and Social Care
9	Rose Gray	Policy Manager	Cancer Research
10	Louise Coleman	Policy Manager	Genetic Alliance
11	Sandy Starr	Communications Manager	Progress Educational Trust
12	Ann Dalton	Consultant Clinical Director	Sheffield Children's NHS Foundation Trust
13	Shahid Hanif	Consultant	Association British Pharmaceutical Industry
14	Fiona Maleady-Crowe	Head of Ethics	Genomics England
15	Simon Wilde	Communications Manager	Genomics England
16	Dr Wendy Inglis Humphrey	Project Manager, Scottish Genomes Partnership	Centre for Genomic and Experimental Medicine, University of Edinburgh

## 1.5 Interpretation of findings

As noted in the executive summary, applying criteria used in the social science literature<sup>22</sup> to determine the credibility of qualitative research findings, we can be confident that the principles and views presented here are credible and valid due to the following strategies used in this dialogue: accounting for bias, meticulous record keeping and systematic analysis, validation and data triangulation. The culmination of this public dialogue is this report which provides detailed and nuanced evidence on how citizens' views, concerns and aspirations can be operationalised in a genomics future.

For reporting on dialogue we use the conventions of qualitative social science reporting:

- We indicate via "a few" or "a limited number" to reflect views which were mentioned infrequently and "many" or "most" when views are more frequently expressed. We use "some" to reflect views which were mentioned some of the time, or occasionally. Any proportions used in our reporting should be considered indicative, rather than exact.
- However, we also indicate strength of feeling even when views are expressed by a minority, as this may also give useful insight into the range of feelings which exist within different groups of people.
- We are reporting perceptions rather than facts; in the case of this project there are various misconceptions our participants expressed about questions of fact, for example lack of understanding of legislation around data protection, and low awareness of biomedical research and its technical processes. We have indicated where we are reporting perceptions of participants, and where we are offering analysis of the implications of these perceptions.
- Where views apply only to a subset of participants, e.g. participants in London, we highlight this in the text.

### 1.5.1 Stylistic conventions

We have used the convention of describing the word data in the singular rather than plural, plus the terminology around patient data recommended by Understanding Patient Data<sup>23</sup> (e.g. 'data or information about you' rather than 'health information'; talking about individual care rather than clinical care; and describing data as either personally identifiable or depersonalised).

<sup>22</sup> <https://ebn.bmj.com/content/18/2/34>

<sup>23</sup> <http://understandingpatientdata.org.uk/>



## 2 The social contract in healthcare today

### 2.1 Public views of key behaviours in the social contract now

The NHS Constitution is founded on a common set of principles and values that bind together patients, the public and staff in order to ensure that it can be effective and equitable. It recognises that each party has important rights that must be respected, but also that each owes each other responsibilities.

Through this combination of reciprocal rights and obligations the NHS aims to operate fairly and effectively for mutual benefit. **The NHS Constitution is thus the expression of a form of 'social contract'** which aims to bring the highest levels of human knowledge and skill to save lives and improve health.<sup>24</sup>

The dialogue participants were not familiar with the term 'social contract' or the explicit concept to which it refers. However, when dialogue participants discussed how they think healthcare works today, and how genomics should operate in society, through the dialogue, they underpinned their responses with assumptions about a clear **social contract** that they believe is in place. They saw it including three principles; **reciprocity, altruism, and solidarity**.

Each principle is important to participants. They framed discussions of healthcare in sometimes contradictory ways, emphasising transactions on some occasions (the principle of reciprocity) and generosity on others (the principle of altruism), and the provision of care based on need, rather than citizenship (the principle of solidarity), overall these three themes were always present, and participants felt they were necessary features of the social contract.

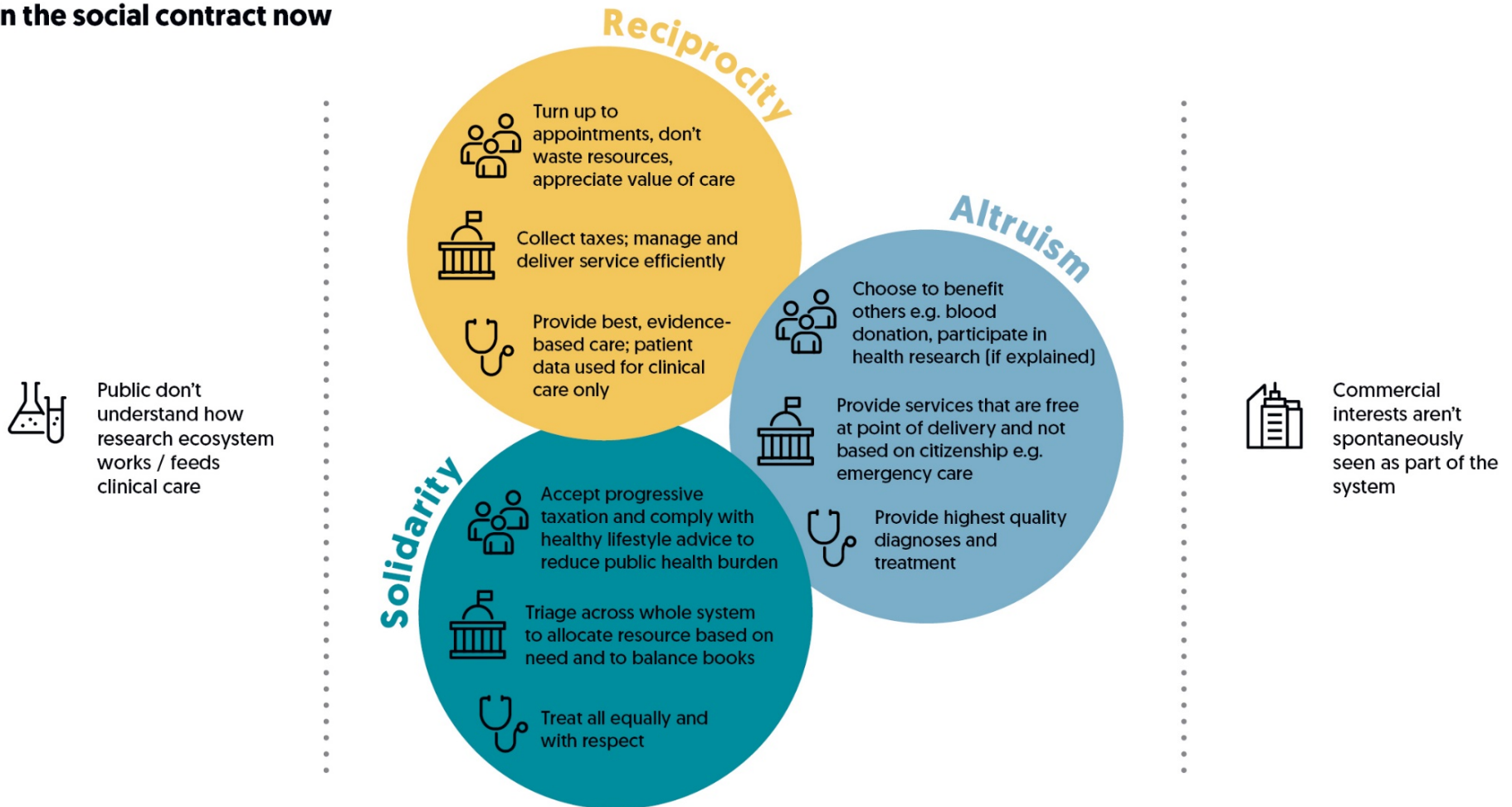
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24

[http://discovery.ucl.ac.uk/1561587/1/Montgomery\\_Ethics%20and%20the%20social%20contract%20for%20genomics%20in%20the%20NHS.pdf](http://discovery.ucl.ac.uk/1561587/1/Montgomery_Ethics%20and%20the%20social%20contract%20for%20genomics%20in%20the%20NHS.pdf)

Figure 2.1: Public views of key behaviours in the social contract

**Public views of key behaviours in the social contract now**



## 2.1.2 Reciprocity

There is a large part of the social contract in healthcare which is seen as essentially **transactional**. In this transaction, the public pay through direct taxation which represents what they give to the system.

In terms of what users get, participants expected the NHS to **provide evidence-based care**, and to treat and help the patients who come through the door.

*“People will come in, and are dealt with when they come in, they will be seen when they have their appointment time.”* London event 1

Patients are **given diagnoses and information** about their condition and the **chance to discuss** it with their doctor.

Participants also felt that **part of reciprocity was the public having high expectations of care**, wanting the most effective treatments possible irrespective of the cost, and treatments that benefit but don't do harm (i.e. side effects).

*“Diseases that have not been eradicated today...Cancer is the biggest one.”* Leeds event 1

*“I think there will be better medicines available. Cures for diseases we can't currently cure. Hopefully cure for cancer at some stage, the common cold.”* Coventry Event 1

When we came to discussing genomics, participants also expressed high expectations for rapid and tangible advances in healthcare; in Chapter 6 we discuss the implications of this and how expectations might need to be managed carefully.

In the reciprocal healthcare relationship, the clinician is trusted to **share patient information and data only with those who are directly involved in a patient's care**. Participants expected that the standard in today's healthcare was that clinicians would otherwise keep patient information confidential.

*“I just thought NHS medical records would stay in the NHS, with your doctor, because of your confidentiality.”* Coventry event 1

In this transactional relationship, participants thought that the **Government's role is to manage and deliver healthcare services efficiently, through taxes collected centrally**.

However, the reciprocal relationship is also seen as **under strain due to the demand** of patients for higher quality care, an ageing society, a rise in population levels, problems in mental health, as well as in the social care system with its knock-on effect on the health service were all mentioned spontaneously. Participants really were worried that the NHS may not be able to fulfil its part of the deal going forward.

*“Everybody's worried. It seems people are more worried about the NHS than optimistic about the future of the NHS.”* London event 1

*“I think the services need to be joined up. I'm talking about social care and healthcare. A lot of the time, the amount of waste that goes into healthcare in regards to bed blocking, you're talking tens of millions of pounds per year.”* Coventry event 1

Participants overall felt the NHS isn't **sufficiently funded by the Government**, (even noting that the dialogue workshops took place shortly after it was announced that the NHS in England will receive a funding increase totalling £20.5 billion in real terms by 2023/24).

*"The lack of funding to me is a political choice. The UK spends much less of their GDP compared to other countries, and that's a political choice. Our taxes are relatively low. If you want a good NHS you need to raise taxes."* London event 1

And, it was clear that unless there is more confidence in the ability of the NHS to deliver core services then that will impact support for, and buy-in to, genomics.

Some participants felt the terms of the reciprocal relationship were already changing, and spoke about a greater emphasis on flexibility, and personalisation, where the use of data and technology enables services to be delivered more efficiently, and empowers people to make informed choices about how they live their lives.

*"Because of that, we might end up with a situation where we have more flexible and more improved services that involve outreach, so you don't have to go to the hospital setting. The technology will be used so you can do some of it remotely and take the pressure off."* Coventry event 1

As noted in the executive summary, consent is one of four areas of medical practice which the Generation Genome report argues needs re-thinking in the world of genomics. The public are clear that **consent to use data for health research purposes is a core part of the principle of reciprocity**, but participants in this dialogue took this so much for granted that it was unconsciously accepted, and was not spontaneously mentioned. Section 4.1.2 discusses changing expectations of consent.

In terms of the expectations of users of the service, participants felt their role in the reciprocal social contract was **to behave well** (to not abuse NHS staff), to **not waste resources** (turn up for an appointment) and **be appreciative of the care being** provided. This went further than simply appreciating your own care, into expressing support for the NHS more widely.

### 2.1.3 Altruism

However, the **'give and get' transactional mentality is not the only way that participants conceived of the healthcare relationship**.

Participants felt that a working social contract requires people to want to benefit others as well as themselves. Individual behaviours such as blood and organ donation and giving to cancer research were mentioned by many.

*"Things like giving blood have gotten better., and they even say whether your unit of blood was used in this hospital."* Edinburgh event 2

(Once explained) **participation in health research was seen as an altruistic act**. A recurrent theme throughout the dialogue was participants' willingness to support, and participate, in health research, once they heard about it; and even if it didn't deliver direct individual benefits.

The NHS was seen to carry the legal, and ethical duty of providing healthcare services that are **free at the point of delivery**. It was generally accepted that **nobody should be turned away from the NHS**, regardless of a person having UK citizenship.

Although stretched resources are a current political flashpoint, and there were a few mentions of health tourism, participants overall took the **moral stance that it would be wrong to deny care**, especially emergency care.

“It has to remain available. Whichever way it goes forward, it has to be available on the basis of need and not the ability to pay.” Summit

“The good things are quick access, like getting seen straight away in A&E. The service is good, staff have always tried their best with the resources that they have. Coventry event 1

#### 2.1.4 Solidarity

Solidarity was the third principle of the social contract. While reciprocity considered the payment of taxes as a direct payment for care received, the solidarity attitude also acknowledges that we should all contribute to **healthcare through progressive taxation**.

In the dialogue this emerged as a strong social norm; participants valued a healthcare system that is not based on an ability to pay, and which does not require private health insurance. Spontaneously, some expressed willingness to pay more tax, with some calling for a hypothecated tax to ensure the NHS can cope with the demands it faces.

“I can see us drifting towards an Americanised system where it’s going to be attached to insurance.” Coventry event 1

A system based on solidarity, participants thought, involved us all accepting that individual good health contributes to the public good, and complying with health instructions (e.g. vaccinations) to reduce the public health burden.

“You are responsible for yourself as an individual. It’s in your interest to promote your good health. I’m not suggesting you wouldn’t be treated. If you abuse and live a lifestyle that is self-harming, you’re a burden on the NHS and everybody else, so it’s down to education and lifestyle.” Summit

The solidarity elements of healthcare were those which focused on public health; with the aim of making people healthier, improving people’s quality of life in old age, and preventing illness, all of which were seen as making cost savings for all of us in the longer-term.

“Preventative medicine rather than curing. I’m thinking about the money it is going to save later on.” Edinburgh event 1

**Triage was an implicit part of solidarity**; participants wanted services to be directed to where the need is greatest.

“One thing I love about the NHS, the NHS hate postcode lotteries and one of the aims is they can say, ‘It needs to be the same’.” London event 2

Clinicians were seen as having a **moral and ethical duty to treat everyone equally and with respect** – the expectation is that they are non-judgemental, making decisions which are based on a person’s clinical need.

## 2.2 Missing elements: private sector

While participants were able to clearly articulate their expectations of healthcare, and why they value it, there was an overall **limited understanding of how the healthcare system works**.

There was an implicit **assumption that the system which drives healthcare is run and managed by public sector decision makers**, who are motivated by the public interest, rather than the private sector, which participants think of as always motivated by profit.

**“Companies don’t do anything for nothing.”** Leeds event 1

Because the **healthcare system isn’t sufficiently understood by its users** (the dialogue participants), there was initial consternation among some about private companies being involved in healthcare.

Among some this was based on a belief that private sector interests should not be allowed to have any involvement in public institutions like the NHS, as ‘private’ was associated with a decline in service standards.

Once explained that the NHS has for many years partnered with commercial companies to ensure the NHS can provide services, and equipment (e.g. electrocardiogram) that meet the needs of its users, the importance of public-private sector partnerships was acknowledged. But, there remained an **unease among many about companies profiteering from people’s vulnerability and poor health**, as they perceived it.

### 2.3 Missing elements: research ecosystem and how research feeds clinical practice

Participants did not mention researchers spontaneously, (neither based in public nor private sector), having little prior knowledge of the role of research in healthcare<sup>25</sup>. The dialogue participants were largely unaware of the framework of research which underpins and pushes forward medical advances.

Overall, participants felt that **striving for more effective treatments was part of the reciprocal responsibility of the healthcare system**. However, this constant improvement to care is, of course, only possible if based on a thriving research sector.

As **health research is something which many haven’t considered**, few understood the distinction between basic and translational research – at best there was a vague appreciation of late stage clinical trials – or the cost of innovation.

**“I don’t know what they [pharmaceutical companies] do. Do they focus on innovation and technology? I think it’s unrealistic that the NHS can fund that explicitly.”** Summit.

There was a limited understanding of how research is funded, the range of organisations undertaking research, or, of why new medical discoveries will increasingly emerge from collaboration. A few even suggested that the NHS should “just go it alone and make medicine itself”.

That said, there were some individuals (from a higher socio-economic group) who knew more about the health research ecosystem, the cost of innovation, and appreciated that commercial companies are integral to improvements in clinical care.

**“They are linked. The research, the universities, hospitals, it’s a whole. They are all working together. They’re the ones who are providing money, investing. The NHS benefits too.”** London event 2

**“You can’t paint drug companies as an evil system. Without the research, there wouldn’t be any progress or cures for treatment. It’s an interdependent system. It has to go through massive amounts of research.”** Summit

Finally, because **clinical research was seen as separate from research practice**, participants also didn’t appreciate how clinicians and researchers are bound by different codes of practice and ethics, which

<sup>25</sup> This lack of knowledge about the framework of research which underpins healthcare, and the role of the private sector in healthcare, chimes with findings from similar dialogues we have conducted such as the public dialogue for [AMS on public views of data-driven technologies](#); and the public dialogue for the [HRA/HTA on consent to medical research](#)

means the nuance of understanding the clinical / research differences and overlaps in the context of genomics was difficult for participants to grasp. We discuss this issue further in the next chapter.



## 3 Public views of what genomics will add to the social contract

### 3.1 Overall views of genomics and the participants' journey

In order to put into context public views of what genomics will add to the social contract, we first need to set out the public's starting points, and how their views were shaped during the hours they spent discussing genomics. Participants formed their views based on experts' remarks, prompts from the facilitators with very detailed discussion guides (see annex), and stimulus presentations from a range of experts. Across the events participants gained views from other participants, bringing up issues, sharing their opinions, and asking the experts for clarification and having long and in-depth conversations that the rest of the group could hear. Ipsos MORI have drawn our conclusions on the different participant journeys that each of them took. This is summarised in section 3.11 below.

#### 3.1.1 Limited understanding of genomics but lots of optimism once explained

**We know that the public have a limited understanding of genomics.** Indeed polling we did for Genomics England found that two-thirds of the public know very little or nothing at all about genomics<sup>26</sup>. In the dialogue discussions almost nobody had heard of a 'genome' or 'genomics', which chimes with empirical research commissioned by Genomics England that showed 85 per cent of British adults had not heard of the word 'genomics' before<sup>27</sup>.

A few participants in this study had however bought a genetic testing kit from 23andMe and Ancestry.Com; and in section 7.1 we suggest that a better understanding of why people are willing to share genetic data in such a context could be useful insight for decision-makers seeking to encourage genomic donation.

There was good recognition of the term 'genes', and lots of **interest in how they influence a person's health** relative to a person's diet, lifestyle and area where they live; and, (once explained) some felt that genetic information was an **opportunity to "nudge" people to make better choices**, that improves public health.

As just over half of people (53 per cent) think that the UK Government does too little to help people live healthy lives<sup>28</sup>, **genomics seems to present an opportunity for the UK Government to be seen as enabling more healthy lifestyles.**

Common genetic disorders like Down's syndrome and cystic fibrosis were well known, but conditions affecting population groups were less familiar e.g. sickle cell disease.

**The accuracy and reliability of genomic analysis** was a worry, particularly to the dialogue participants from a Black and Minority Ethnic (BME) background, given historically poor access to health services and underrepresentation in clinical trials. Therefore, the public expect that researchers and clinicians

<sup>26</sup> National representative polling Ipsos MORI did for Genomics England in May 2018 found that two-thirds of the public know not a lot or nothing at all about genomics / genomic medicine. The full report and topline findings can be accessed at the following link:

<https://www.ipsos.com/ipsos-mori/en-uk/public-awareness-and-knowledge-genomics-genomic-medicines-low>

<sup>27</sup> Middleton A. Socialising the genome: making genomics resonate. *F1000Research* 2018, 7:149 (slides). Detailed findings can be accessed at the following link: <https://doi.org/10.7490/f1000research.1115249.1>

<sup>28</sup> <https://www.ipsos.com/ipsos-mori/en-uk/public-and-nhs-70-years-together>

must inform patients about the boundaries of the genetic test such as its familial implications, and its uncertainties such as its probabilistic nature, and do this in an intelligible way.

Also a particular worry for BME participants, as well as other participants, was that a **genomics data database or 'library'** as experts in some of the dialogue workshops described the idea of a genomic database, **creates a possibility for genomic data to be used by the authorities to create a 'surveillance society'**. All participants said emphatically that this would fracture trust in data donation, and was considered a red line. Section 4.3 further discusses participants' red lines.

Overall, however, in our dialogue there was a **very positive response to genomics**. Special benefits were seen for cancer, rare and ultra-rare diseases, long term life limiting illness, the lives of children, and improvement to patient choice with more knowledge about individual predispositions.

*"The whole thing makes me feel quite optimistic. Something like dementia, taking strides to something like that is worthwhile. Joined up thinking and working together is a great thing."* Leeds event 1

However, the public have **high (somewhat unrealistic) expectations**, with many participants envisaging a near-term future with new treatments and personalisation of care, and significant cost savings for the NHS.

*"The amount of money that will be saved for the NHS by the prevention of these conditions, rather than treatment, has to be beneficial."* Summit

As such, in the short term public expectations will need to be carefully managed (see Chapter 6); the information the public will need in the first few years after launch is likely to be different from information needs ten years into the programme. Changes in the wider world may also condition expectations (an example is the recent public debate on a gene editing ban following evidence of human gene editing trials taking place in China.)

After hours discussing genomics, **almost all were relaxed about their health and genomic data being used in health research**. This support though is conditional on consent (section 4), the use of de-identified data as routine and red lines being respected (section 5), robust risk assessment and safeguards being implemented and maintained by policy makers (section 7), and of course, clinical impact.

*"That's what changed with me from the first session. When we were told your data is shared, I was like, 'Oh my god.' As the discussion went on, I realised the importance of sharing data, it shows up trends."* Coventry event 2

#### The views of participants in the Edinburgh workshop<sup>29</sup>

The views expressed by participants in the Edinburgh workshop were effectively the same as those in the workshops in England. This chimes with what we know already about attitudes to sharing patient data for health research which is that a nexus of educational attainment, socio-economic group and digital literacy, rather than geography, are more influential in shaping people's views.<sup>30</sup>

<sup>29</sup> As agreed with Genomics England and the Scottish Genome Partnership we have analysed the Edinburgh workshop results together with the results from the dialogue workshops conducted in England. The report contains quotes from all of the events.

<sup>30</sup> Ipsos MORI (2016) Public attitudes to commercial access to health data pp. 85 <https://wellcome.ac.uk/sites/default/files/public-attitudes-to-commercial-access-to-health-data-wellcome-mar16.pdf>

Overall, the Edinburgh participants were relaxed about developing genomic testing further in NHS Scotland and expected research findings to lead to new ways of working and improvements in care.

### 3.1.2 Issues in genomics were initially seen as similar to others in healthcare

In general, many of the issues around genomics initially raised by participants are similar to others in healthcare, and although the language and specifics may be new, many of the ethical and practical issues are seen as the same. These include:

- **Doctor taking responsibility** – the clinician is still seen as responsible for providing the best care possible. However, the dialogue participants were worried that stretched resources in the NHS will mean that the system won't be able to cope, and the benefits of genomics won't be equally redistributed in the longer term.
- **Evidence-based decision-making** – participants want doctors and clinicians to be empowered to make their own choices, and are worried about doctors becoming 'bogged down' in administrative justification for their decisions. Moreover, participants expect that decisions made by clinicians are informed by the best available scientific evidence, and it was felt that genomics would enable better decision making, and access to new treatments.
- **Incidental findings** – participants weighed up a number of factors when discussing if they would want to be told about incidental findings which are not derived from genetic analysis: a positive prognosis of the 'other' condition, a person's emotional resilience, and support to deal with the consequence of knowing the result. These factors were the same in genomics, although once they reflected on familial implications, this added a complicated ethical dimension to their decision-making (see section 4.1.1).

## 3.2 Public views of what genomics will add to the social contract

### 3.2.1 The same core principles, but new intersections between them

The rest of this page is a brief overview of the key points which are presented in the remainder of Section 3.

By the end of the dialogue, participants had discussed the implications of genomics for the social contract. Overall, in a genomics-driven healthcare of the future, they felt the principles of **reciprocity, altruism and solidarity** would need to remain core to the social contract, and require largely the same behaviours of all the actors as they do at the moment.

For example, clinicians are expected to treat all equally and with respect, and patients turn up to appointments and appreciate value of care.

However, the new science of genomics means that the **three elements might intersect in new ways** - leading to **new expectations of behaviours of all actors in the system**.

**Reciprocity intersects with altruism:** Genomics requires donation of data from individuals. This blurs the line between reciprocity and altruism. The public in the dialogue saw giving their data as an altruistic act - but also expected benefits would probably accrue to their families and descendants.

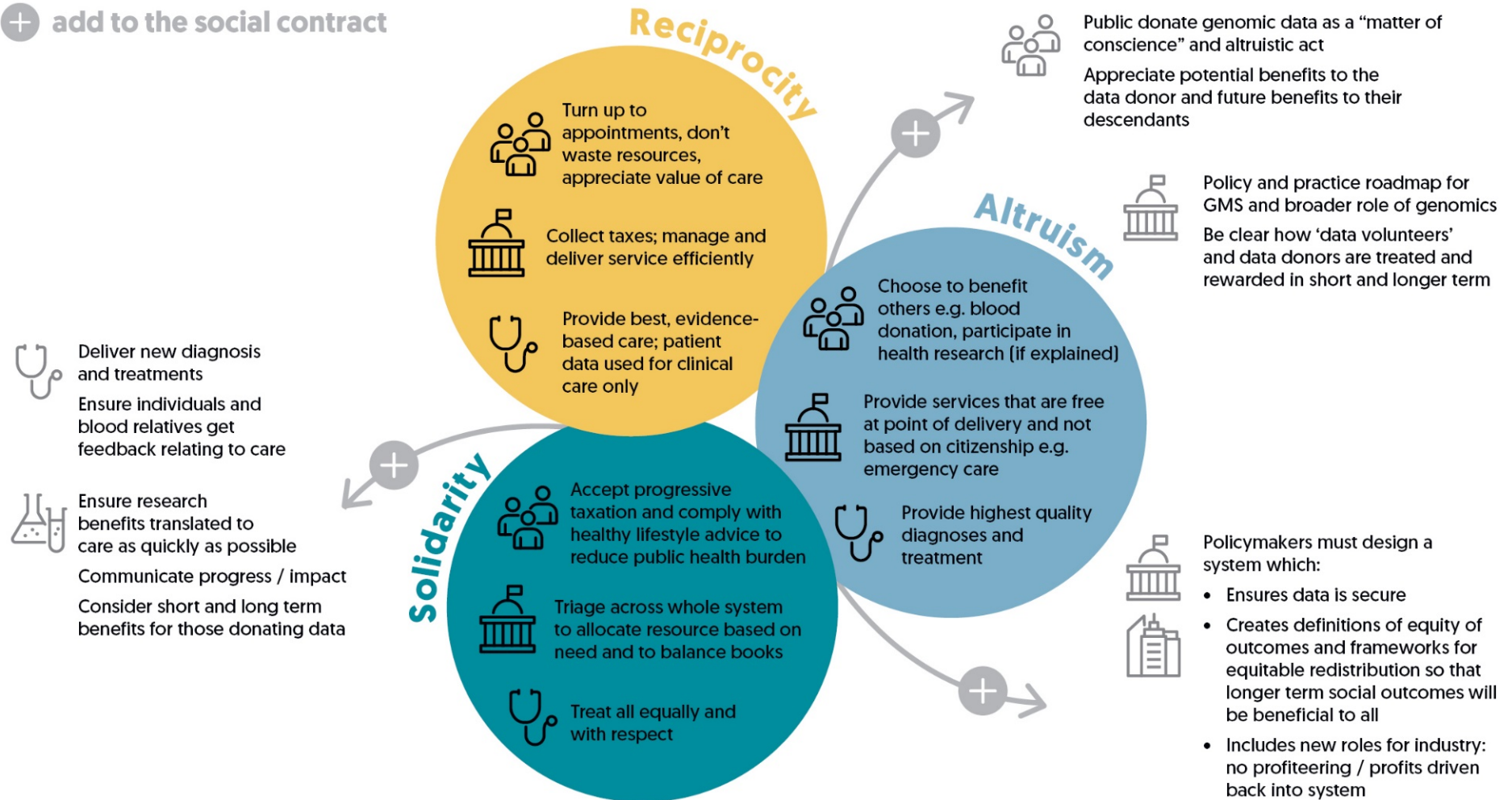
Similarly, **reciprocity and solidarity also blur**. Genomics can use patient data in research and clinical care at the same time, creating a feedback loop between both to increase possible learning. Participants felt that this **research/clinical blurring** brought the ideas of reciprocity and solidarity closer together.

**Solidarity and altruism** also become more closely connected. In the future, if large genomic dataset(s) exist, and can potentially be accessed by new actors, (such as commercial companies) social solidarity will be affected by the altruistic acts of individual donors.

Figure 3.1: Public views of what genomics will add to the social contract

## Public views of what genomics will

+ add to the social contract



### 3.2.2 Reciprocity links with altruism

Participants quickly **grasped that genomics requires donation of data from individuals**, and when experts described how they hope genomics will impact healthcare, they saw **giving their data as an altruistic act**. This blurs the line between reciprocity and altruism.

*"It's part of your social contract to make your information available. Your genomic information might hold the key to a breakthrough in research."* Edinburgh event 2

Initially it was thought that when people give their genomic data, they will get more back than they do currently, but **during discussion they came to realise that benefits would probably accrue to their families and descendants**.

*"It could mean the gift you pass on to your family. If there are any problems in the future which are health-related, they could be helped."* Summit

Therefore participants thought there would be an increased expectation that the public would donate data, although it was clear that **data donation should not be mandatory**.

*"I think genomics is brilliant. Collecting the data for the next generation, they need it. That's a wonderful thing to be able to do that."* Coventry event 1

**Participants saw the genomic medicine service as altruistic**, in that they felt the learning and insight derived from those who participate in the service should be used in the treatment of other cancer and rare disease patients who are offered whole genome sequencing or genetic testing, in order to see how their situation compares.

Participants were happy in principle to support those who chose not to donate data, but who would still be treated; and they felt that nobody should be forced to give their genomic data, instead people need **educating about the benefits of altruism** (genomic donation), and the **benefits of being altruistic**.

*"It's about, so far, one of the biggest things coming out of this is sharing the data, so if they get the foundations right, once you've gained the trust, it's about educating the public so they tick the box to share."* Coventry event 2

This was because they could recognise, albeit after hours of discussion, that genomic medicine relates to us all – comparing all our genomes must be seen to be done in an altruistic way, with a concern for the wellbeing of all. This is seen by participants as particularly intrinsic to genomics, **making altruism even more important than it has been in the past**.

*"It's an investment into the future. My grandchildren might benefit."* Coventry event 2

*"You have to keep yourself informed, so that it's an everyday thing that we have a conversation on. Knowledge is power. We're tuned into the message of having a flu jab every year. We need a message as strong as that, so that we all know what's happening."* Coventry event 2

As well as feeling altruistically that genomics is a human-scale project and all in society can ultimately benefit from its advances, there was a **feeling that treatments derived from genomics should benefit those contributing to the system**, and people in the UK especially.



Participants thought therefore that government and healthcare policy makers would need to create a roadmap for genomics where the status of data donors is made clear, and any rewards for them carefully worked out, to balance the interests of altruism and reciprocity.

### 3.2.3 Solidarity links with reciprocity

Solidarity in genomics is predicated on an assumption of collective action; that all agree to take part in, to bring about improvements in clinical care and health research.

Genomics can use **patient data in research and clinical care at the same time**, creating a feedback loop between both to increase possible learning. Participants felt that this **research/clinical blurring** brought the ideas of reciprocity and solidarity closer together. This gives researchers and clinicians **new duties of care**.

A changing relationship between clinicians and patients is already apparent as a result of uptake in new types of data gathering (e.g. wearables), and the use of technology in delivering services (e.g. virtual appointments).

But, given the way research can feed clinical treatment and vice versa, **participants were clear that researchers have new responsibilities in genomics**.

First, participants felt that both clinicians and researchers in a genomics future would be responsible for ensuring that patients and families are informed about the progress of research.

Second, both researchers and clinicians should also focus on ensuring the translational benefits of research to care, to benefit all of society, as quickly as possible.

**“Potentially faster diagnosis of problems. That would lead onto much quicker decisions about what treatments are best suited to that particular individual rather than a spectrum of stuff.”** Leeds event 2

Third, both researchers and clinicians should communicate, what they are doing with genomic data, and the link between their work and eventual discoveries with clinical impact; so the public can know their collective action is worthwhile and having impact.

Finally, the public think that patients are in a unique position to help shape the service that takes shape around them – and therefore clinicians (and researchers) should engage and take their views on board as much as possible.

### 3.2.4 Solidarity links with altruism

In the future, if large genomic datasets exist, and can potentially be accessed by new actors (such as commercial companies), participants felt social solidarity will be affected by the altruistic acts of individual donors.

**“Genetics feels like an individual thing, but it’s very social. The results always implicate other people.”** Coventry event 2

Dialogue participants felt that policy makers should take a long-term view and design a system which prevents dystopian social outcomes in the future.

Participants emphatically **did not want their donation of genomic data used to create a stratified society** which disenfranchises vulnerable members. (e.g. genomic datasets being heavily used in insurance, marketing, etc – which are discussed in Chapter 5 below).



There would be a **new role for industry** in bringing the benefits of genomics to society; but either **voluntary or external regulation will be needed**. Again, participants did not want to see the altruistic data donations of individuals used to enable private sector profiteering (discussed in Chapter 5).

Ultimately, the public want policy makers to design a system that prevents these things from happening in practice. **Rational ignorance**<sup>31</sup> means participants are willing to defer its design to the NHS and experts with no vested interests (such as a panel of “lay-experts” e.g. 100,000 Genomes Project Participant Panel<sup>32</sup>). Section 7 discusses what the governance framework of this system should look like.

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<sup>31</sup> It's impossible to know everything. Some degree of ignorance is inevitable, and we all must personally decide what is worth knowing and understanding. **Rational ignorance** helps each of us decide what information would be most useful. Rational ignorance means intentionally choosing to remain uninformed on a topic because the cost of acquiring the information is greater than the estimated potential benefits. <https://study.com/academy/lesson/rational-ignorance-definition-effect.html>

<sup>32</sup> <https://www.genomicsengland.co.uk/participant-panel/>

## 4 What the public think will change in the social contract

### 4.1 Changed expectations around data donation

As well as the additions to the social contract, participants also felt there would be one change to the social contract; **genomics changes the expectation that clinicians will only share data with those involved directly in patient care.**

#### 4.1.1 Changed expectations of confidentiality

Registered medical professionals have both a (legal) duty of care and a (legal) duty of confidentiality to their patients – breaching either of which can be grounds for de-registration. This is recognised to be the fundamental basis of trust in healthcare, and has been for longer than the NHS has existed.

The fact that a genetic result has implications for an individual patient as well as their family members **introduced the dialogue participants to some complicated ethical questions.**

These questions were introduced using hypothetical family tree scenarios (see annex B). For example, participants were told about a woman who finds out she has the BRCA1 gene alteration after having her genome sequenced, thus increasing her risk of developing breast cancer.

They were also told that the woman has a twin sister, but wants to keep the test result between herself and her clinician. The main question was: does the twin sister have a right to know about her sister's gene alteration given the possible implications for the sister's health; and if so, how should she be told?

The resultant discussions identified some barriers to sharing genetic information. **Individual rights were seen as being in tension with family rights** and views could become very polarised and split.

**Spontaneously most participants didn't want data used beyond a person's care**, because confidentiality has always been part of healthcare.

**"If people breach their own confidentiality by their own means, they need to take responsibility. The NHS must keep it secret. If that woman doesn't want to be tested, that needs to be kept secret. If her family subsequently find out from her, that's her responsibility."** Edinburgh event 2

But after discussing the implications of the genetic test result based on the hypothetical family tree-scenario, most did not see **patient confidentiality as axiomatic to the social contract in the context of genomics involving data with family implications.**

**"If the parent's got a potentially serious disease and it's potentially through [knowledge from a test their child's had], the doctor has a duty to tell his parents."** London event 2

They also felt that there was a moral obligation for patients to find out what they can about their own health and, with guidance from genomic counsellors and clinicians pass on as much information to benefit their families rather than harming them by omission by not telling them critical information.

But, after hearing genetic clinicians recount stories of how these implications can play out in their work, some cooled on this idea because they thought it was too idealistic. They felt that some patients and families of patients motivated in a petty or personal way would withhold information of critical importance.

By the end of the discussion, it was felt that there **should be an assumption that patients and their blood relatives will be informed of all medically relevant information.**

**“In this scenario the GP (of one patient) should talk to the GP of the relative.”** Edinburgh event 2

Participants wanted to **empower clinicians to make the final decisions on disclosing information**; but at the same time were concerned not to add any administrative burden on clinicians from a medico-legal perspective.

In order to facilitate this, clinicians need reassurance from policy makers, and from the medical profession so they feel confident to make such decisions, and potentially to develop a transparent framework for how they exercise their expert judgement, so that individuals are not left vulnerable and open to criticism.

**“Legislation needs to be there to protect the practitioner and the public. What would that look like? They can sort that in the Houses of Parliament.”** Coventry event 2

Finally, there was a feeling among some (who tended to be participants from a high social economic grade) that genomics and the new era of data-driven technology in healthcare will usher in a social contract characterised by clinicians who **understand, actively manage, and communicate risks and tensions to patients.**

#### 4.1.2 Changing expectations of consent

There is a significant amount of public opinion research on consent for use and access of patient in health research. Indeed, [our recent dialogue for HRA/HTA<sup>33</sup>](#) tested the public’s comprehension and acceptability of broad consent, so-called hybrid consent (100,000 Genomes Project consent form), and dynamic consent which allows participants to adapt their consent preferences.

The focus of this study was to uncover the nuances of consent in the context of genomic data, rather than repeat the consistent and clear research that already exists.

Spontaneously participants in this dialogue wanted **assurances that consent was built into the process of genomics.**

**“You’ve got to give your consent, a right to refuse.”** London event 2

Initially, the dialogue participants thought that consent should make it explicitly clear who will have access to their data and how it would be used, but after it was explained it is not always possible to be so clear upfront, there was a pragmatic agreement that **prior consent may no longer be possible for all uses.**

And so participants accepted that there may be un-knowable outcomes from genomic research, and as a result they are willing to let go of knowing precisely which research projects will use their data, for the sake of personal and social benefit.

**“There should be an opt in system. If I had the option today, I would be diving right in there.”** Edinburgh event 2

But, they were very clear that doesn’t mean their willingness to participate in research is unconditional, as consent must be based on research which has been ethically-approved and is for the public good.

<sup>33</sup> [https://www.hra.nhs.uk/documents/1570/Consent to use human tissue and linked health data in health research FINAL.pdf](https://www.hra.nhs.uk/documents/1570/Consent%20to%20use%20human%20tissue%20and%20linked%20health%20data%20in%20health%20research%20FINAL.pdf)

**“As a nation, because genomics is a good, everyone should be opted into it. If you want to be opted out, you can.”** Coventry event 1

The focus of this dialogue was not to define what constitutes “ethically-approved”, and rational ignorance means participants are willing to defer safeguards and governance to both the NHS and experts who have no vested interest in genomic research.

The dialogue participants were also clear that the public should have their dissent to their genomic data being used in health research recognised<sup>34</sup>, and have a choice about those who have access to the data (e.g. commercial interests). Almost nobody felt they would opt out, as they could accept that an increased risk to privacy is worth the diagnoses and personalised medicine they might receive.

#### 4.1.3 A new need to inform and educate the public about what has changed

Participants felt both clinicians and researchers should be equipped with **“genomic literacy”** to support the informational, emotional and practical needs of patients and data donors; giving more information and support than has been necessary in the past when communicating with research participants.

Participants suggested that clinicians in particular should seek out genomic literacy opportunities so they feel confident discussing the uncertainty of diagnosis / treatment, and are able to convey information derived from genomic analysis to lay people.

**“If I were told by a genetics counsellor that I had a condition that affects my family, I would have no children.”** Coventry event 2

The dialogue reveals areas where communication skills might be important. There was **limited understanding of the nature of a genetic test result**, and even after information from experts, some participants spoke about it in terms of a binary yes / no answer i.e. certainty. This will need to be explained carefully to patients and data donors in future.

Once the nature of the test result was explained, views about feedback were often based on perceptions of certainty of diagnosis, risk of harm, likelihood of getting it, or time between the test result and on onset of symptoms.

When these nuances were discussed, participants wanted Government to ensure the NHS has the resource it needs to deliver excellence in communication throughout the “genomics pathway”: genomics counsellor, clinician, researcher.

**“The most important thing is to ensure that the relevant structures are in place to make it work. We’re talking about people like counsellors, otherwise it won’t work. Lots of projects have lots of money put into them and 8 years down the line, the plug gets pulled.”** Coventry event 2

**“These people must be specifically trained and that will cost money and time. It is a great thing, but huge.”** Summit

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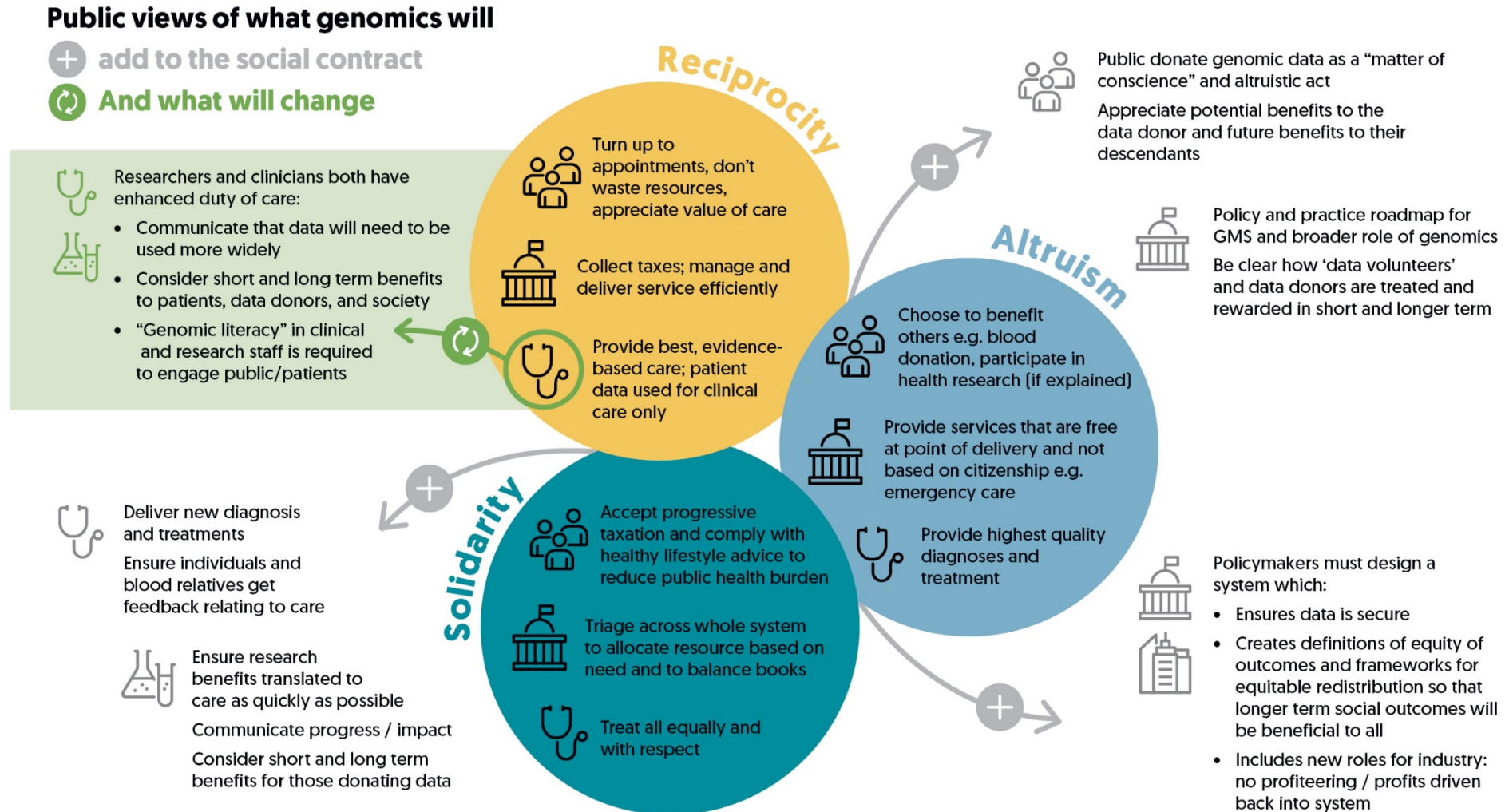
<sup>34</sup> The national data opt-out (NDOP) was introduced on 25 May 2018, enabling patients to opt out from the use of their data for research or planning purposes, in line with the recommendations of the National Data Guardian in her Review of Data Security, Consent and Opt-Outs.

It was acknowledged that informing patients about the meaning of different consent options and the implications of different test results were both very important.

Participants were surprised and reassured when it was explained that genomic counsellors can spend up to forty minutes discussing such things, as they had envisaged an appointment time of around ten minutes.

But they were soon worried that the NHS would not have sufficient numbers of genomic counsellors to meet demand. Assurances around workforce planning and training more genomic counsellors did not convince. They believed these things will take a long time to realise, and in the meantime, they believed there would be long waiting times and geographically patchy services. As such, **policy makers need to make it clear how the system is able to cope with the broader roll out of genomic medicine**, and specifically ensure there are sufficient numbers of genomic counsellors, and clinicians that are literate in genomics.

Figure 4.1: Public views of what genomics will add to the social contract, and what will change





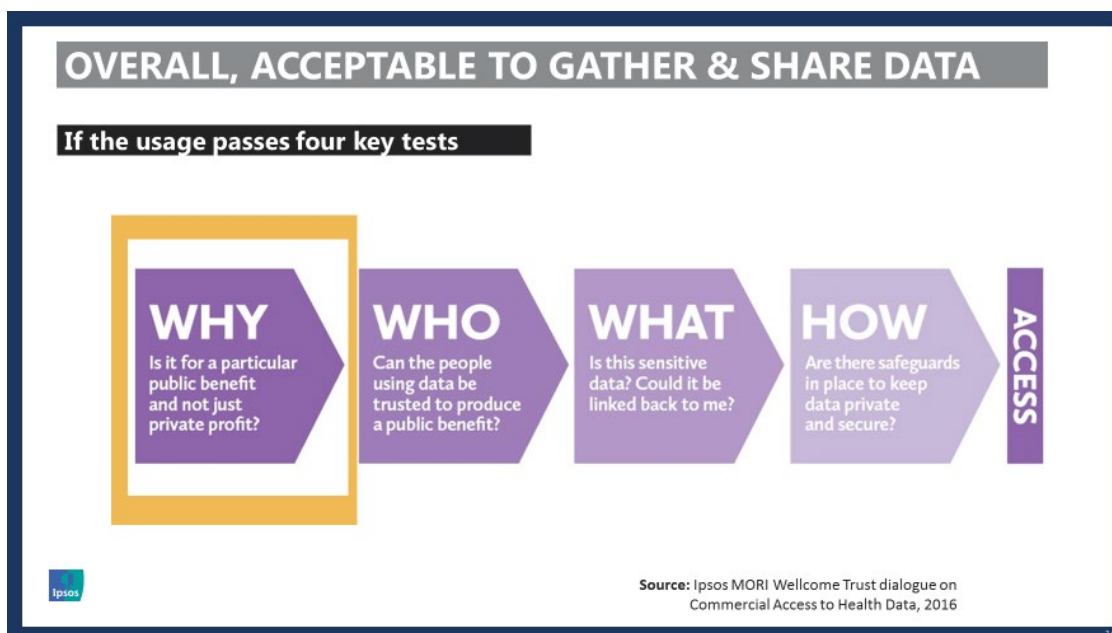
# 5 Public views of genomic data: access and use outside of healthcare

## 5.1 Views of genomic data and access

Reactions to the idea of genomic data being different were less about inherent difference – it was often seen as on a par with other forms of health data – and more about uses of genomic data, particularly outside of healthcare, and the fact that it has familial implications, as discussed in 4.1.1 above.

There was low understanding of the concepts of data aggregation, anonymisation vs de-identified and data linkage; and no spontaneous sense of ‘mutuality’ of data – the fact we can learn more about all of us by looking at the data together. However, when this idea was introduced in the context of healthcare and genomics, people supported it.

Participants’ attitudes to access of genomic data were the same as identified in previous research: in general, the wider public is happy for the NHS to use people’s health data<sup>35</sup>, but there is less support for commercial companies having access, even for research<sup>36</sup>. But overall the public think it is acceptable to gather data, and use it outside the NHS, if the usage passes four key tests identified in our [Wellcome study on commercial access to healthcare data](#)<sup>37</sup>.



<sup>35</sup> Ipsos MORI / Deloitte LLP (2017) The State of the State 2017-18: Austerity, government spending, social care and data. Full survey results can be accessed at the following link: <https://www.slideshare.net/IpsosMORI/the-state-of-the-state-20172018>

<sup>36</sup> Ipsos MORI / The Health Foundation (2018) NHS at 70: public perceptions. Full survey results can be accessed at the following link: <https://www.ipsos.com/sites/default/files/2018-07/public-perceptions-of-the-nhs.pdf>

<sup>37</sup> Ipsos MORI / Wellcome Trust public attitudes to commercial access to health data 2016 <https://wellcome.ac.uk/sites/default/files/public-attitudes-to-commercial-access-to-health-data-wellcome-mar16.pdf>

Participants in this dialogue expressed their views in accordance with these priorities, in comments such as the below:-

**“If it benefits people, I don’t care who the NHS are sharing my data with as long as it’s not Facebook and Google.”** London event 1

Sharing genomic data (with researchers) internationally was an initial red line, as participants assumed standards of data security and governance are not as high as in the UK. Views quickly shifted to acceptance after it was explained that the NHS has put in place all the necessary (statutory) protections, because participants support the idea that genomics will be more quickly realised by sharing genomic information with a community of genomic experts.

**“Bringing it together anonymously would advance knowledge in certain diseases...Why work separately when you could have the great scientists of the world working collectively?”** Coventry event 1

## 5.2 Genomic data use outside of healthcare: insurance

When applying for insurance a person has to disclose any symptoms they experience or any diagnoses, screening, or treatments they receive, if this information is requested on the application form. In some cases, genomic or genetic testing can discover a predisposition to disease.

The dialogue participants were asked to consider how they felt about sharing a genetic test result if they were asked to disclose this information.

It is worth briefly setting out the context in which reactions to this question are formed. **The insurance industry has some trust issues:** In 2017, only around a quarter of British adults said they trusted the industry<sup>38</sup>.

The **use of genetic test results in insurance was initially a big shock**. There was lots of high feeling, and some felt so strongly about sharing data with insurers that they said they would not donate their genomic data.

**“I’d be less inclined to have my genome sequenced if I knew it would affect my insurance.”** Coventry event 2

There was **almost no recognition of mutuality**: the key principle of commercial insurance, whereby individuals pay according to the best estimate of the risk that they bring with them; or the *uberrima fides* principle, or utmost good faith, where each side declares all it knows about the risk<sup>39</sup>. The insurance industry was **associated with ‘sharp practices’ when it comes to honouring insurance claims**.

**The starting point was often that insurers should not have access to additional health data**. In terms of genomic data, it was felt that it would enable insurance companies to extract maximum premiums from people, punish people for poor health, and make insurance unaffordable to those who need it most.

There was **almost no recognition of the importance of pricing risk accurately**, and that this is good for the health of society, as well as commercial sustainability.

<sup>38</sup> Ipsos Global Trends Survey, 2017. Full survey results can be accessed at the following link: <https://www.ipsosglobaltrends.com/>

<sup>39</sup> <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1691992/>

And, when it was suggested that premiums might go down if a test result showed a low health risk, the dialogue participants just didn't believe it would happen.

**"It's like they want to know whether you're a good investment or not, in a sick twisted way. I don't know. I'm not sure."** London event 2

Participants were told that a new code of practice for insurers had just been issued<sup>40</sup>. It was explained that it is not a binding law but a code to which the industry signs up that is taken very seriously by the companies involved i.e. the members of the Association of British Insurers.

It was **hard for participants in this dialogue to grasp the voluntary nature of the code of conduct**, which chimes with how participants in our HRT/HTA dialogue on consent felt about the moratorium for sharing genomic data with the industry<sup>41</sup>. In both dialogues, **participants were worried that it would be difficult to enforce, and hard for consumers to hold companies to account.**

Figure 5.1: dialogue stimulus of HMG/ABI code of practice for insurers

### Code of Practice for Insurers

- An insurer will not require or pressure an applicant to undertake a predictive or diagnostic genetic test in order to obtain insurance.
- The results of a predictive genetic test may be considered in an application for insurance only when both of the following conditions are met:
  1. This Code states that the specific predictive genetic test may be considered and;
  2. The sum assured exceeds the financial limits set out in this Code.

And, **there was consternation about the parameters of the code (see figure 5.1, and figure 5.2 overleaf)**. The inclusion of Huntington's disease – the only genetic condition which insurers are permitted to ask applicants to disclose – was even questioned, unless it has been substantiated that the applicant has inherited the defective gene.

<sup>40</sup> A voluntary code of practice agreed between HM Government and the Association of British Insurers on the role of genetic testing in insurance was published by HM Government and Association of British Insurance in October 2018. It can be accessed at the following link: [https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment\\_data/file/751230/code-on-genetic-testing-and-insurance.pdf](https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/751230/code-on-genetic-testing-and-insurance.pdf)

<sup>41</sup> [https://www.hra.nhs.uk/documents/1570/Consent to use human tissue and linked health data in health research FINAL.pdf](https://www.hra.nhs.uk/documents/1570/Consent%20to%20use%20human%20tissue%20and%20linked%20health%20data%20in%20health%20research%20FINAL.pdf)

Figure 5.2: dialogue stimulus of HMG/ABI code of practice for insurers

Type of insurance	Financial limits above which predictive genetic tests may become relevant	Medical conditions for which insurers may ask for and take account of predictive test results, for policies above the financial limits
Life Insurance	£500,000 (per person)	Huntington's disease
Critical Illness Insurance	£300,000 (per person)	None
Income Protection Insurance	£30,000 per annum (per person)	None
All other types of insurance	Predictive genetic test results will not be asked for, or taken into account, whatever the level of cover.	

"Some diseases affect certain groups. Breast cancer affects women more. You're going to start discriminating against population groups." London event 2

Ultimately, participants thought the best they could hope for was that policy makers would carefully monitor compliance with the code, because they were resigned to insurance companies adding more conditions each time the code was reviewed.

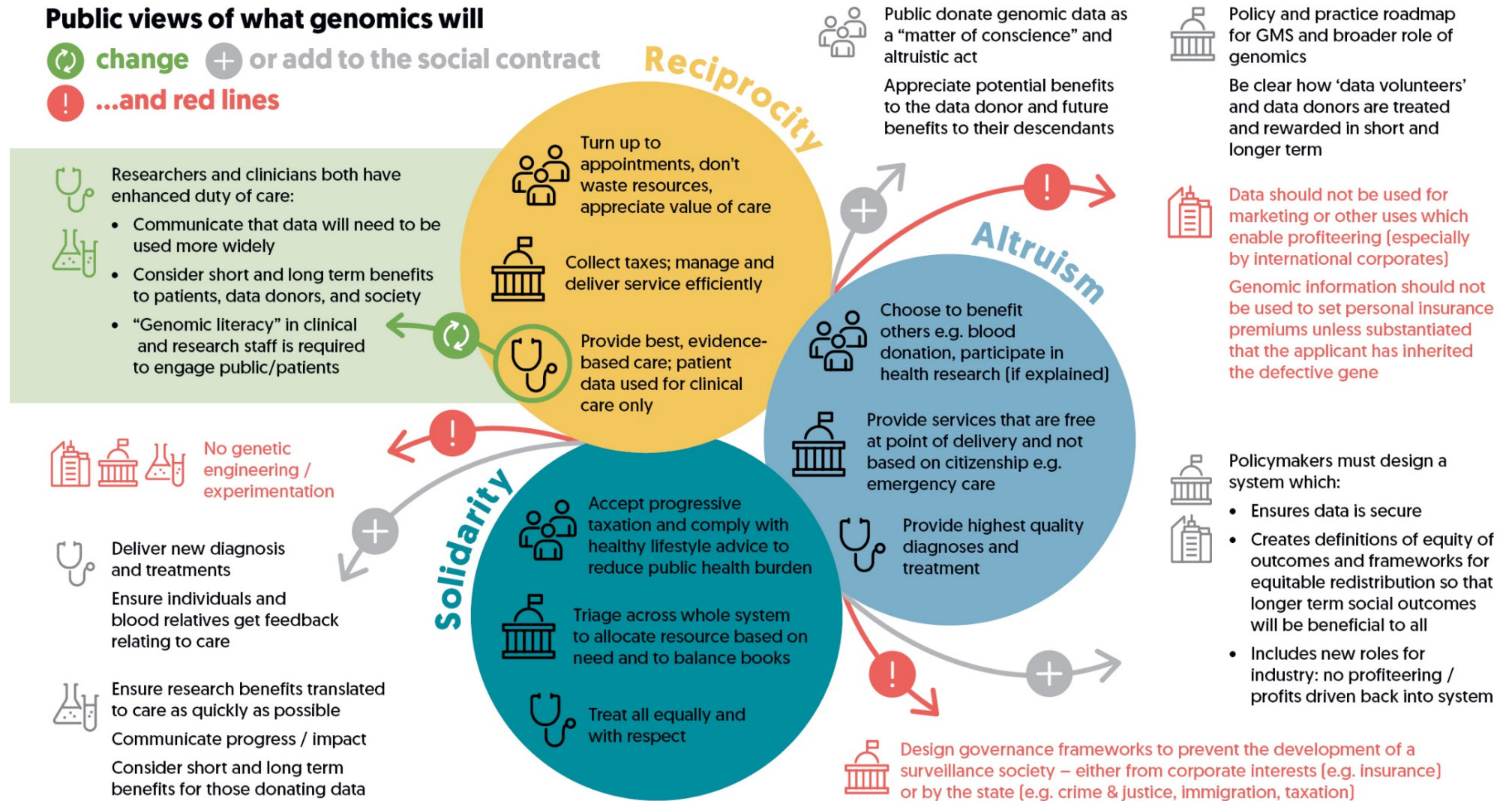
"What's stopping the insurance companies when they get their foot in the door, not making it wider? They could lower the life insurance amount and make more illnesses available." Coventry event 2

### 5.3 Genomic data use outside of healthcare: the public's red lines

Assurances around the de-identified nature of the data meant participants are less concerned about personal harms.

However, participants are concerned about broader societal harms, and they identified some clear red lines and boundaries when it comes to how far genomic data and information derived from genetic analysis should be used outside clinical care and health research, as shown in figure 5.3.

Figure 5.3: Public views of what genomics will add to the social contract, and what will change... and red lines





### 5.3.2 Genetic engineering

Participants were concerned about the possible **unintended consequences arising from the field of genetics** (rather than an NHS Genomic Medicine Service); in the extreme, enhancing human capabilities, and eugenics, were mentioned.

**“Another bad point is that we might be becoming too clever with things like genetic engineering. It’s an ethical problem.”** Edinburgh event 2

**Genetically edited babies** was seen as the natural next step, and assurances from experts that “it [gene editing] just won’t happen in the GMS” were insufficient, as there was a feeling this would happen by ‘accident’, rather by design.

**“A ‘must not do’ is genetically modifying babies in the future. Designer babies.”** London event 2

When experts explained the difference between genetic modification where there is a gene known to cause a debilitating illness (e.g. Huntington’s disease) and changing a feature of a child (e.g. gender, eye colour, hair colour), almost all supported the idea of improving the wellness of a child by modifying their genetic make-up.

A few, however, in particular participants whose child has a genetic condition such as Down’s syndrome saw advances in genomics leading to **more aborted pregnancies**, and more **prejudice and discrimination for people with a genetic condition** (e.g. Turner Syndrome<sup>42</sup>) than there is in today’s society. Most though, when discussing the fact that fetal screening test for Down’s is currently available, concluded the idea of a more accurate prediction would benefit some parents.

### 5.3.3 Surveillance society

There was real concern that collecting genomic data creates a possibility for it to be used to monitor and track citizens, either by corporate interests (e.g. insurance) or by the state (e.g. crime and justice and taxation).

**“It should stay within the NHS. It shouldn’t be used for things like ancestry.com where they were used by criminal services. It’s crossing the boundary of privacy. It should be kept in the research field definitely.”** Edinburgh event 2

The idea of a repository of DNA data was spontaneously associated with a perception of institutional racism, racial bias in stop and search practices, and the so-called hostile environment immigration policy. Although this was a clear red line, there was a sense of fatalism that the day would come when genomic data would be used to monitor and, discriminate against minority and disenfranchised groups in society.

**“Not sharing with the police either. If you’ve got my DNA, you don’t have the rights to share that with the police.”** London event 2

**“They’ll be picking which country to deport you back to which you have no link to whatsoever. That’s my worry.”** London event 2

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<sup>42</sup> <https://www.nhs.uk/conditions/turner-syndrome/>



It was felt that insurance companies should not be permitted to use genomics tests for predictive tests. While there was interest in the accuracy of prediction in healthcare, in insurance it was considered a red line as participants believed that analysis would be used to identify people who are likely to have high personal insurance premiums, and as a result reduce access to much needed protection.

#### 5.3.4 Administrative and political uses

Although nobody made an explicit connection with combining genomic data and administrative<sup>43</sup> data, there was a general feeling that more data is being collected all the time which has the potential to be used in ways which people did not consent to or would not vote for: cutting choice, reducing access to services such as welfare, and a sense of control of UK citizens were all mentioned as clear red lines.

#### 5.3.5 Targeted marketing

Genomic data is seen as potentially very valuable to marketers, and participants were very clear that data should not be used for marketing or other areas which enable profiteering, especially by international corporates.

There was a feeling that pharmaceutical companies and insurance companies would link genomic data with other data outside healthcare, in order to target individuals who are more susceptible to these approaches due to their vulnerability (e.g. health condition/illness).

*"I'd be worried about drug companies finding out you have certain illnesses and then they target you for testing and certain medications. Say my data was known and I was shown to get an illness, then I'd be targeted by drug companies for research. I might not want to address my illness, I might not want to think about it, rather than being targeted and reminded. It affects your mental health."*

Coventry event 2

*"Once they've [insurance companies] got that information, they can use that for their marketing. They always have access to it."* London event 2

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<sup>43</sup> Administrative data collections are built up by government departments and agencies during their day-to-day activities. They routinely gather information, when registering people or carrying out transactions, or for record keeping – usually when delivering a service. Such data can include: social security payment records; educational attainment records; health records, court records; and tax records.

## 6 Communicating genomics

### 6.1 Genomics communication context

Raising the public's awareness of genomics is already a priority for policy makers. Just a few examples are: the Department for Health and Social Care, and its partners NHS England and NHS Scotland announcing the achievement of sequencing 100,000 genomes<sup>44</sup>, the UK Government announcing an ambition to analyse five million genomes in the UK over the next five years, plus the publicity around DNA testing for children with cancer in NHS England.<sup>45</sup>

Alongside these efforts, academics, social scientists and science communicators are involved in public engagement projects which seek to raise public awareness of genomics, as well as build support for it. For example, the Socialising the Genome<sup>46</sup> initiative funded by Wellcome and Genomics England looks at what sort of narratives and metaphors raise awareness and seeks to encourage more people to talk about genomics<sup>47</sup>.

However, this study (including the literature review) and empirical evidence from Ipsos MORI<sup>48</sup>, shows that a lot **more work is needed to raise public awareness, and understanding, of genomics.**

Before setting out how this can be achieved, it is worth briefly considering **the context in which genomics is happening. Now is a risky time for data usage.** Ipsos's global survey for the World Economic Forum found widespread distrust on personal data usage by companies and governments, and as shown in the graphic below, most in society don't know what data organisations and companies hold about them or what they are doing with it<sup>49</sup>.

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<sup>44</sup> To deliver the 100,000 Genomes Project, NHS England established 13 NHS Genomic Medicine Centres (NHS GMCs) responsible for recruiting and consenting patients, providing samples from both blood and cancer tissue and the clinical information for analysis; validating results; and working with patients and clinical staff to broaden the use of genomics across clinical specialities.

<sup>45</sup> <https://www.bbc.co.uk/news/health-46777387>

<sup>46</sup> <https://www.genomicsengland.co.uk/socialising-the-genome/>

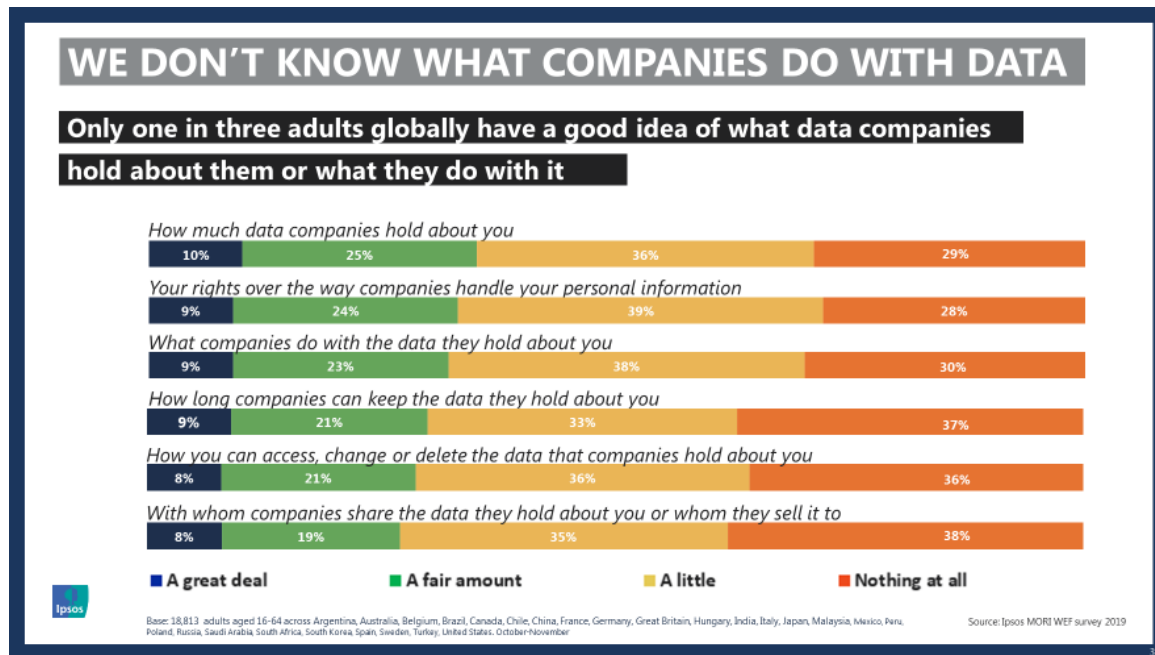
<sup>47</sup> A short preliminary overview of some of the findings can be accessed at the following link:

[https://www.thelancet.com/journals/lancet/article/PIIS0140-6736\(17\)31011-5/fulltext](https://www.thelancet.com/journals/lancet/article/PIIS0140-6736(17)31011-5/fulltext)

<sup>48</sup> National representative polling Ipsos MORI conducted for Genomics England in May 2018 found that two-thirds of the public know not a lot or nothing at all about genomics / genomic medicine. The full report and topline findings can be accessed at the following link:

<https://www.ipsos.com/ipsos-mori/en-uk/public-awareness-and-knowledge-genomics-genomic-medicines-low>

<sup>49</sup> <https://www.ipsos.com/ipsos-mori/en-uk/global-survey-reveals-widespread-distrust-personal-data-usage-companies-and-governments>



At the same time, the UK faces some significant challenges: inequality and poverty, an ageing society, and the NHS remains the public's biggest worry after Brexit.<sup>50</sup> As such, there is a lot of 'background noise' that genomics messaging will need to cut through.

In the remainder of this Section we offer our ideas on how best to communicate genomics and the broader roll out of genomic medicine. They are based on the explicit views of participants in this dialogue, as well as our analysis of the kinds of messages and phraseology that resonated, plus other relevant studies which have looked at ways to inform the public about sharing their health data.

## 6.2 Genomics framed as a collective endeavour

Realising the potential of genomics will require a **critical mass of UK citizens supporting it to the extent they are willing to participate** – while this is a longer-term aim, creating and implementing a genomics narrative should be an immediate priority – the dialogue participants were clear that policy makers and the NHS have a responsibility to do more to educate people about genomics.

The Socialising the Genome initiative has stated that actors who work in genomics have different ways of talking about it, so there is also a need for a **shared narrative expressed in intelligible language** which is used by all actors in genomics.

The dialogue participants were clear that the NHS will need to manage public expectations of genomics in the NHS. Therefore, the narrative will need careful framing, because, if the benefits of genomics are over-hyped, or the uncertainties downplayed, there is a **risk of fracturing the public's trust in genomics**, and health research more generally.

**“You make a conversation about it. It becomes a part of your consciousness. If it just gets rolled out, people aren't going to be comfortable with it.” Summit**

We suggest that communications should convey the idea that genomics is an **ambitious, ground-breaking programme of activities** that will:

<sup>50</sup> <https://www.ipsos.com/ipsos-mori/en-uk/issues-index-august-2018-public-concern-about-eu-and-brex-it-remains-historically-high-levels>

- re-define what best care is: for example genomics is moving away from “one size fits all” treatments to patient care tailored to their unique genetic make-up
- deliver new medical discoveries
- but also change healthcare in ways which cannot be predicted with certainty.

This framing will resonate with the dialogue participants’ optimism in genomics, while adding a degree of realism that it’s still very early stage and lots of learning and insight still awaits.

“I think being transparent as well, like not ignoring potential risks and making people really excited about the benefits.” London event 2

The participants from the 100,000 Genomes Project who engaged with dialogue participants introduced a sense of realism as to what genomics is able to deliver, and helped the public engage with the idea of uncertainty in genomics. As such, we suggest these personal contacts could be useful in wider communications. Examples could also help manage public expectations – for instance, talking about cystic fibrosis, which was one of the first conditions to have its genomic component properly understood and yet no genomic treatment has arrived in the NHS in over 30 years.

The genomics narrative should not be confused with a service like blood or organ donation, which was a key misunderstanding in the dialogue discussions. Instead, communicating the idea of a “**national moonshot**” or shared endeavour may work to link genomics to a wider UK programme of innovation in which we are all involved.

We suggest messaging **shouldn’t focus on the technology and details of the science or the clinical and research divide**, because these things are too complicated to take on board through direct messaging, and in any case these technical details were not required by the dialogue participant to understand the broader issues such as the uncertainty of diagnosis / treatment, the probabilistic nature of a genetic test result, and the fact it has familial implications

Instead, we recommend that the NHS explain the **paradox in genomics**: that an individual’s data has little to add to understanding health whereas data submitted by a crowd of people is much more likely to yield insights. Only by building and analysing a ‘**genomics library**’ will patients get individual benefits.

This is an important aspect of any communications as it helps people engage with the idea of the benefits of altruism and altruistic benefits, and speaks to a common worry that researchers are spending their time looking at just one individual’s patient record.

There was interest in how / whether other countries are looking to mainstream genomics, and the fact that the UK is a trailblazer struck a chord with participants. **Genomics can be framed as one of the UK’s success stories**, although this should not be the primary message as healthcare, rather than economic growth, appears to resonate more.

“We seem to be quite far behind. Are we catching up here? If America has already introduced [genomics] into healthcare, where are we?” Coventry event 2

Policy makers should tell the public that **genomics benefits won’t be realised without a partnership** between the NHS, research community and commercial companies. This needs to be explicit given the knee-jerk reactions to private sector involvement in healthcare. There is therefore a need for a

description of public and private partnerships in layman's terms, and a crystal-clear explanation of how genomics is funded (who is paying for what?), and whether and how benefits are reinvested.

### 6.3 A mission statement for genomics

In the longer-term, genomics will need a call to action - if the new social contract is going to work the public will need to know what genomics is and be motivated to take part, so there is a need for a mission statement that conveys the idea that success needs solidarity, altruism and reciprocity. We can infer from the participants' discussions, and expert input, with regard to the social contract in genomics that the following statement may work as a call to action, as well as communicate the essence of the social contract in an intelligible way.

**"People powered genomics".**

As genomics evolves, further public and stakeholder engagement may be needed to ensure the social contract remains relevant and instructive.

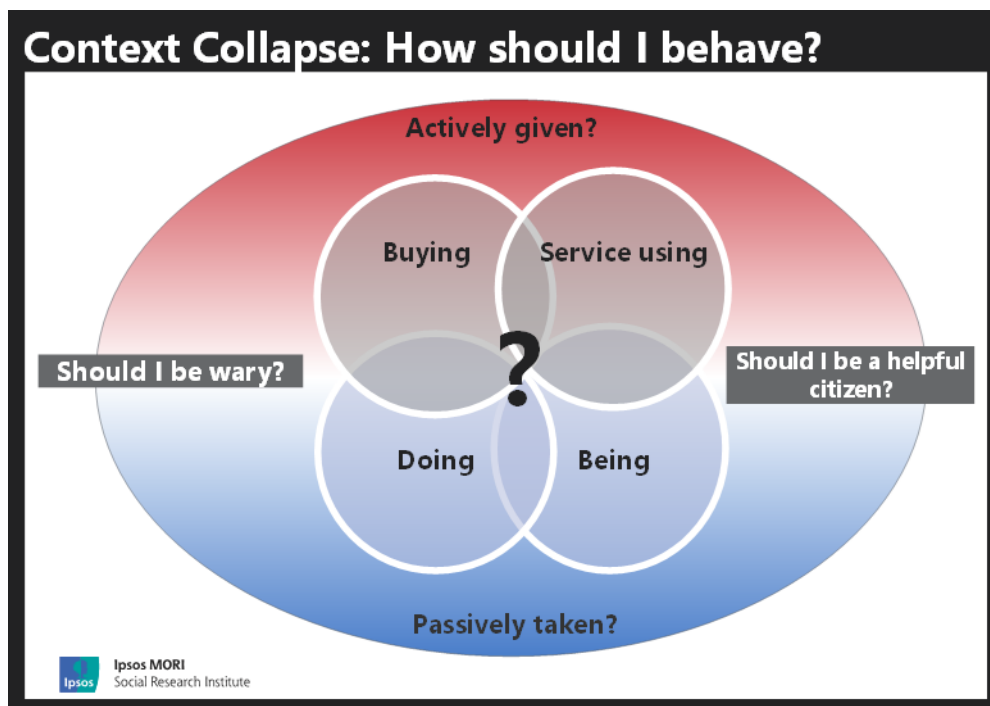
## 7 Conclusions

### 7.1 Next steps from this dialogue

This project has built on all the work done already which has covered a range of topics related to genomics including ethics, law, social contract, consent, NHS capability, and funding; it has looked at attitudes to health data and data sharing among the public, and population sub groups e.g. BME, and stakeholders. This insight, plus the input from experts from a range of private, public and charity organisations, as well as contributions from the public and patients have enabled us to offer conclusions and advice for what to do next in the NHS Genomic Medicine Service.

However, there are areas where we would recommend further investigation, in particular around the intersection of genomic medicine and research, and the sharing of health data with commercial companies.

The Wellcome commercial access to health data project, mentioned throughout this report, identified that **attitudes to data sharing are influenced by the context in which it is used**. Overall, we found that people are relaxed about sharing their data with the NHS because they have an expectation that their data is being used on their behalf. When sharing data consciously in a commercial context, customers give their non-health related data, because they get a service they value in return. When it comes to **sharing health data with commercial companies** however, these contexts collapse, and people find it hard to know what to expect, and can become uncertain<sup>51</sup>.



In that study we did not specifically explore genomics – but some commercial genomic services currently blur the boundaries between ‘commercial’ data sharing, where there is a more wary customer relationship, and ‘public sector’ data sharing, where there is an assumption that the

<sup>51</sup> <https://wellcome.ac.uk/sites/default/files/public-attitudes-to-commercial-access-to-health-data-wellcome-mar16.pdf>



patient's best interests are considered. Some participants in this study bought a genetic testing kit from 23andMe and Ancestry.Com, but didn't consider how their genetic data would be used or the possible repercussions of knowing their test result.

We noted that the approach they used to finding out about their genome could be described as a '**gamified consumer relationship**'. In these relationships, people appear less concerned about giving information about themselves when it is seen as "quirky", a "curiosity" or a "bit of fun". We would recommend researching this context further, to see how far people understand the risks and benefits to themselves. If people give their data to a consumer testing company and something happens that fractures trust in genetic testing, this could have implications for trust in genomic medicine.

## 7.2 Governance framework

This dialogue also gives us learning on public views of a governance system for genomic data in future.

There was healthy scepticism about how well any data sharing system can ever work in the NHS given that it is not known for high tech data management. As one participant said:

**"It's like a dinosaur, it's not ready to handle the technology". London event 2**

Therefore, **policy makers and the UK government will need to provide reassurance on the use of patients' data**. Specifically, they will need to inform the public that they will remain responsible for patient data and ensure the system conforms to the highest standards of data security and governance across sectors and national jurisdictions, and be accountable (individually and organisationally) if things go wrong.

Researchers only being allowed to access de-identified data if it remains on NHS servers was a key reassurance, however there is the possibility that rules around data sharing may evolve in the future. Therefore, it will **be prudent for those managing and delivering the genomics system to make the case for data sharing in genomics and health research more generally**: that a global effort by a community of experts will help translate researching findings to in clinical care more quickly.

In the longer-term, there may be need for a **wider narrative about the changing nature of data usage** by the NHS and its partners, specifically what the NHS and researchers are doing with donated data, which will need to take place on a national level.

The public understood that a genomic database cannot be 100% secure, and that any database can be hacked; while this dialogue didn't explore attitudes to standards and safeguards (e.g. national security standards of data protection), it was felt they need to be of the highest standard.

Finally, **access to data by insurance companies, or other private sector organisations, needs to be carefully reviewed**, as the new ABI code approach was seen as too 'voluntary', leaving too much to the discretion of private companies.

## 7.3 New definition for equity of outcomes and equitable redistribution

The literature review and this dialogue clearly show that the public, patients, and experts are all concerned about the current and future capabilities of the NHS, and it is clear that the extent to which

the public will give their support for genomics will be based on whether it is seen to be delivering better clinical outcomes.

In the short-term, the dialogue participants recognised that genomics is in its early stages and **could accept that the benefits of genomics won't be equally distributed across the UK population**. They appreciated that creating more effective treatments for cancers and rare diseases are very difficult problems, and appreciated that the GMS is seeking to build on the lessons learned and insight from the 100,000 Genomes Project.

But, in the longer-term, the **public want to be reassured that their altruistic act will not be wasted**, and that the budget for genomic medicine allows all sections of society to benefit fairly. Therefore, policy makers will need to build trust by ensuring that any GMS is properly funded, which means that the NHS (whether in England, Scotland, Wales or Northern Ireland) has the resource it needs to deliver excellence throughout the "genomics pathway": genomics counsellor, clinician, researcher. The long-term roll-out should be **nationally balanced both in terms of equity of access and outcomes**.

The NHS will need to inform the public how much genomics costs in the context of the overall healthcare budget; and give a realistic assessment of cost savings over time. Policy makers and the NHS will need to tell the wider public **what are the expected timescales for adding other conditions and diseases to the genomic test directory**<sup>52</sup>, so as to manage expectations, while maintaining support for investing in genomics.

If the UK public are donating data, this brings in attitudes to the private sector and as such participants thought that the public has a right to expect that some of the financial rewards of new medical developments will return to 'UK plc'. And therefore, participants thought that **policy makers should implement a mechanism** which delivers treatments for rare/ultra-diseases, a fair price for new treatments, and re-investment in clinical care and research, in recognition of the fact that genomics creates new financial opportunities for pharmaceutical companies.

#### 7.4 Inscribing the genomics social contract in the NHS constitution

Actors in the genomics system should not use the term social contract; it is not widely understood and is not likely to resonate with the public. However, participants in this dialogue concluded that the **elements of the social contract do need to be written down**, and then communicated to everyone who has a role to play in genomics.

The key elements which need to be communicated are the **new expectations of behaviours of all actors in the system**, which we have noted elsewhere in this report.

**"There have to be guidelines in the world of genomics. Everyone has to understand where the responsibilities fall."** Leeds event 2

- **Reciprocity ↔ Altruism:** Genomics requires donation of data from individuals. This blurs the line between reciprocity and altruism. The public in the dialogue saw giving their data as an altruistic act - but also expected benefits would probably accrue to their families and descendants.

<sup>52</sup> <https://www.england.nhs.uk/publication/national-genomic-test-directories/>

- There would, therefore, be an increased expectation that the public would donate data, but participants felt that data donation should not be mandatory.
- Participants thought that government and healthcare policy makers would need to create a roadmap for genomics where the status of data donors is made clear, and any rewards for them carefully worked out.
- **Reciprocity ← → Solidarity:** Genomics can use patient data in research and clinical care at the same time, creating a feedback loop between both to increase possible learning. Participants felt that this **research/clinical blurring** brought the ideas of reciprocity and solidarity closer together.
  - Both clinicians and researchers in a genomics future would be responsible for ensuring that patients and families are informed about the progress of research.
  - They should also focus on ensuring the translational benefits of research to care, to benefit all of society, as quickly as possible.
  - Both researchers and clinicians should communicate the progress they are making towards discoveries with real clinical impact; so the public can know their collective action is worthwhile and having impact.
- **Solidarity ← → Altruism:** In the future, if a large genomic dataset exists, and can potentially be accessed by new actors, (such as commercial companies) social solidarity will be affected by the altruistic acts of individual donors. Dialogue participants felt that:
  - Policy makers should take a long-term view – design a future which prevents dystopian social outcomes. Participants emphatically did not want their donation of genomic data used to create a stratified society which disenfranchises vulnerable members. (e.g. genomic datasets being heavily used in insurance, marketing, etc)
  - There should be a new role for industry in bringing the benefits of genomics to society; but either voluntary or external regulation will be needed. Again, participants did not want to see the altruistic data donations of individuals used to enable private sector profiteering.

Given the profound effect that genomics is likely to have on healthcare in the UK, **we recommend inscribing in the NHS constitution these elements above;** the ways in which genomics adds to, and changes, the social contract.

## 8 Annexes

This annex contains the following documents:

- **Annex A:** Event 1: discussion guide, plenary presentation, and stimulus materials
- **Annex B:** Event 2: discussion guide, and stimulus materials
- **Annex C:** The genomics summit: discussion guide

## Annex A

Genomics: time for a new social contract? Public dialogue to uncover views of mainstreaming genomic medicine in UK

*Event 1: 3-hour evening session – 6.15pm-9.15pm.*

Time	Question areas and materials
6.15pm-6.30pm	<p>Arrival, registration, refreshments</p> <p><b>WELCOME AND PLENARY</b>  <u>Lead facilitator show slides 2-4</u></p> <p><b>Slide 2:</b> Study overview: study sponsors i.e. GE and SW, and overarching aim: <b>your thoughts, hopes, aspirations, and concerns about different ways people’s genomic information can be used in NHS.</b> Introduce dialogue delivery team, clients, experts/ observer and</p> <p><b>Slide 3: The main areas to cover</b> – don’t worry you don’t need to have prior knowledge, we will give you all the information you need and experts here who can answer your questions <b>Explain workshops taking place in:</b></p> <ul style="list-style-type: none"> <li>• 4 locations: London, Coventry, Leeds, and Scotland</li> </ul> <p>Event 1: evening            Event 2: day-long, Saturday</p> <ul style="list-style-type: none"> <li>• And a final genomic medicine summit Friday 14<sup>th</sup> Dec.7 people from each location invited back to the summit.</li> </ul> <p><b>Slide 4:</b> housekeeping, plus breaks, toilets and fire alarm</p>

6.30pm-  
7.05pm

## Understanding Healthcare and health data

### TABLES 6.30-6.40pm

*Facilitator introduces themselves, thank you for coming, no right / wrong answers - really interested in what you think.*

*Before we do introductions, I'd like you to write down what you think of when I say the future of healthcare in the UK. Here are some post-its and I want everyone to write down what they think will be "good or better" about healthcare and on another post its what you think will be bad or get worse*

*Okay so now please tell us your name and what you have written down, each ppt shows 2 post-its each. **Facilitator cluster post its on Flipchart***

- IF TIME ASK: What do you hope healthcare will be able to do in the next ten years? *Prompt on - For you, your relatives, patients, people with long term of serious conditions, for different groups like children and the elderly...*

### PLENARY 6.40-6.55pm

#### QUIZ

Split into 2 groups per table i.e. cc. 5 ppts in each group and handout quiz sheet. In your group, discuss then write down what you think is the correct answer.

Facilitator to ask teams to shout out answers.

- Q1 correct answer is a, b, c, d, e. then show **slide 6**.
- Q2 correct answer is a, b, d, e, f, then show **slide 7. Then show slide 8** – healthcare – it's not just one thing.
  - *If needed, say, in your own words, the following extract from Understanding Patient Data: Companies are involved in many ways in the delivery of care and research across the NHS, but there are strict controls on how companies can use patient data, to protect your privacy. The NHS can't do all the data analysis on its own. It has to work in partnerships, with academic researchers, charities, regulators - and commercial companies.*



- Q3 correct answer is a, b, c, e then show *slide 9*. Then say: information from patient records can be used to help *understand more about disease, to develop new treatments, to monitor safety, to plan services and to evaluate NHS policy*.
  - If needed, say, in your own words, the following extract from *Understanding Patient Data* then say, in your own words, the following extract from *Understanding Patient Data* - *The NHS will never share your name or contact details **with companies to use for marketing purposes**, unless you give consent. Pharmaceutical companies have to follow strict rules about marketing, and they are not allowed to advertise prescription medicines to patients in the UK. If you are applying **for life insurance cover**, an insurance company will want to know information about your medical history. The insurer will usually ask you questions about your lifestyle and relevant family history but may also want to see your medical records. This is only possible with your permission. Only if you agree, your GP will then provide the relevant information to the insurer. There is currently a ban on insurers using genetic information*

Lead facilitator show video: Understanding Patient Data - Patient data saves lives: the bigger picture

<https://www.youtube.com/watch?v=fJ2hyXCOOyQ>

#### TABLES 6.55-7.05pm

- Any surprises in what you've just heard? Any questions?
- Response to info on **organisations involved in healthcare**.
- Do you ever think about your health data, or any data about you collected? Did you know about these various kinds of ways data is collected?
- What role will patient data play in the future of healthcare? Probe: what would a positive future look like? And a negative one?

7.05pm-  
7.35pm

**Introducing genome sequencing and 100,000 Genomes Project**

#### PLENARY 7.05-7.15pm

Lead facilitator shows video: Introducing genomic medicine, stop video at 4min30 seconds

<https://www.genomicsengland.co.uk/the-100000-genomes-project/understanding-genomics/>

Facilitator explain that 'genetics' is often used as a catchall word when searching for faults in one gene and 'genomics' is used to explore many genes to find the fault in one gene. They both test the same thing but the genomic test gives you the chance to explore many genes at once, in your search for the single relevant one. The key relevance of 'genomics' being important is that it offers a rich set of data that can be used in all sorts of research.

**Lead facilitator show slide 11**, introducing the idea that genome sequencing has become a lot cheaper and results are known a lot quicker due to advances in "reading" DNA and computational (computer) capabilities, so it's more accessible to people.

#### TABLES 7.15-7.25pm

- How does that sound? Do you like the idea of genomic sequencing being made available to more people? Are there any pros and cons?
- Have you heard of any organisations collecting genetic / genomic data? E.g. 23andMe, AncestryDNA, Genomics England, Illumina etc.
- Can you think what these organisations are collecting genetic / genomic information for? E.g. *People's curiosity in their ancestry, curious about their chances of developing a genetic condition, have an undiagnosed health problem*
- How do you think genomic data compares to the types of patient data health we discussed at the start? It is more / less sensitive / useful than the kind of health information in your medical record?

**Table facilitator show slide 12**, introducing the idea why genetic information can be useful

- What impact do you think collecting more genetic information will have?
  - patient / person / family: *give certainty, or prolong uncertainty, influence decisions affecting lifestyle, life planning,*
  - *increased demand on primary care such as GP / support*
  - *UK more attractive place to do health research / biomedical science.*

#### **PLENARY 7.25-7.35pm**

**Representative from GE show slide 13** introducing the 100K Genome Project and then **14**, introducing the idea why genetic information, and linking it with other health data and lifestyle data can have clinical and research benefits.

	<ul style="list-style-type: none"> <li>• Any surprises in what you've just heard? Any questions?</li> <li>• Are there any pros and cons?</li> </ul>
7.35pm-7.50pm <b>BREAK</b>	
7.50pm-8.15pm	<p><b>Experts with different points of views discuss their hopes for, and concerns about, making genomics / genomic medicine part of routine NHS care</b></p> <p><b>PLENARY</b></p> <p>Depending on numbers at each event, 2 or 3 experts spend a few minutes introducing their area of work / professional background in plain English / layman terms, then explain what they see as benefits and risks What are their hopes for rollout? What they think the key issues are?</p> <p><i>Table facilitators invite experts to talk to ppts / answer their questions.</i></p>
8.15pm-8.50pm	<p><b>Making genomic medicine part of routine NHS care</b></p> <p><b><u>TABLE DISCUSSION 8.15-8.50pm</u></b></p> <p>Okay, we've heard from people who are involved in the 100,000-genome project and genetics services. Now we're going to hear about how the genomics medicine service will be phased into the NHS and I will ask for your thoughts. Again, experts are on hand to answer your questions. So, if everyone looks at:</p> <p><b><u>Who will genomic medicine be made available to first?</u></b></p> <p>slide 16 Facilitator note: key take-aways are: who WGS will be made available to and on what basis and setting the target for how many genomes will be sequenced in the next five years in the context of how many sequenced in the 100k GP.</p> <p><i>Was there anything you didn't understand / expect to hear? Invite questions from ppts and flipchart as will feedback in plenary. Ask why it's important to know and invite experts in to answer questions</i></p> <p>What do you think of what it says? Probe: good, optimistic, bad, worried?</p>

Now let's all look at:

**Making genomic medicine a part of NHS care - data about your genome**

slide 17 Facilitator note: arrangements for data storage security, organisations which will have access to genetic data and for what purposes, process for obtaining access.

Was there anything you didn't understand / expect to hear? *Invite questions from ppts and flipchart as will feedback in plenary. Ask why it's important to know and invite experts in to answer questions*

What do you think of what it says? Probe: good, optimistic, bad, worried?

Is it fair to opt of out of research and planning? What if scientists find out something that could be useful in clinical care?

Any surprises in terms of what organisations can access genetic information? How do you feel about that? Can you think why data might be analysed and interpreted by researchers working in commercial companies and / or around the world? What do you think about the arrangements for data storage and access? Do you need to know they exist or how they work? Why?

Now let's look at:


**making genomic medicine a part of NHS care (3) – your result and your primary care**

slide 18 Facilitator note: anticipated impact of genomic medicine on patient engagement with primary care services, the HCP-patient relationships.


Was there anything you didn't understand / expect to hear? *Invite questions from ppts and flipchart as will feedback in plenary. Ask why it's important to know and invite experts in to answer questions*

What do you think of what it says? Probe: good, optimistic, bad, worried?

	<p>What do you think of the way the genomic test result will be feedback to the patient? What role should other family members have, given the results says something about them too? What impact do you think genomic medicine will have on health care services? How might it affect the relationship between the healthcare professional and the patient?</p>
8.50pm-9.00pm	<p><b>Hope and concerns for genomic medicine in the NHS.</b></p> <p>Okay, so we want you to think about what the experts have told us as well as everything you've heard tonight. Now, we want you to work in groups of 3 and write down on post-it notes your hopes and concerns for genomic medicine in the NHS.</p> <p>No limit on the number of hopes and concerns but do rank your top 3 in each.</p> <p>Facilitator invite 2 or 3 groups to present top 3 and invite others to compare. Then ask all: on balance, are you optimistic or concerned about how genomic information can be used in the NHS? Do you need more information in order to reach a conclusion? IF SO: what?</p>
9.00pm-9.15pm	<p><b>Thanks and close</b></p> <ul style="list-style-type: none"> <li>• Homework task: genomics conversation and online search, if do complete and bring back we will give you an extra £5.</li> <li>• Questionnaire knowledge of and attitudes to genomic medicine in the NHS, used for segmenting groups at next stage</li> <li>• Evaluation questionnaire</li> <li>• Hand out incentive</li> </ul>





This project is funded by:  
Genomics England and Sciencewise,




They are interested in **your** thoughts, aspirations, hopes and concerns about the different ways genetic information can be used in the NHS


<u>You!</u>	<u>Project funders</u>	<u>Experts</u>
<u>Ipsos MORI</u>	<u>Observers</u>	<u>Evaluator</u>

 Ipsos MORI  
Social Research Institute

**What does your involvement mean?**

- Your **understanding of**, and **support for**, genomic medicine
- Your thoughts on a '**deal**' for genome medicine
- How **comfortable you are** with genomic medicine – is anything **unacceptable**?
- Whether you think any **pros and cons** and how these can **be addressed**



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# Healthcare and information about YOU



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5

### What data exists about you?



**NHS care providers**

Share your **personal** data for your care **WITH YOUR PERMISSION**



Name, address, age, weight, lifestyle data

NHS number

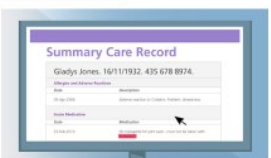
Scan images

Lab results

Letters from the consultant – “free text”

Summary Care Record

Symptoms, treatment, rare conditions



**Summary Care Record**  
Gladys Jones, 10/11/1932, 435 678 8974

**PATIENT IDENTIFIABLE LEVEL**  
Hospital number: 450 257 7104

Medical History	Start date	Follow up
Asthma	July 1986	6 monthly check
Acute back pain	March 1999	
Acute back pain	May 2005	
Acute back pain	December 2010	
Acute back pain	February 2014	
Diabetes Type 2	March 2013	6 monthly check

**Current Prescriptions:**


Verapamil tablet	
Escitalopram	
Methocarbonyl	
Allegiant	
Paracetamol	
Coartem	
Wegovy	
Sanofi	
Aspirin/Chewy	

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### What organisations does the NHS share patient data with?


**NHS care providers:**

- Clinicians
- Health commissioners
- Hospital trusts
- Contractors




**Academic users:**

- Research centres in Universities
- Independent researchers

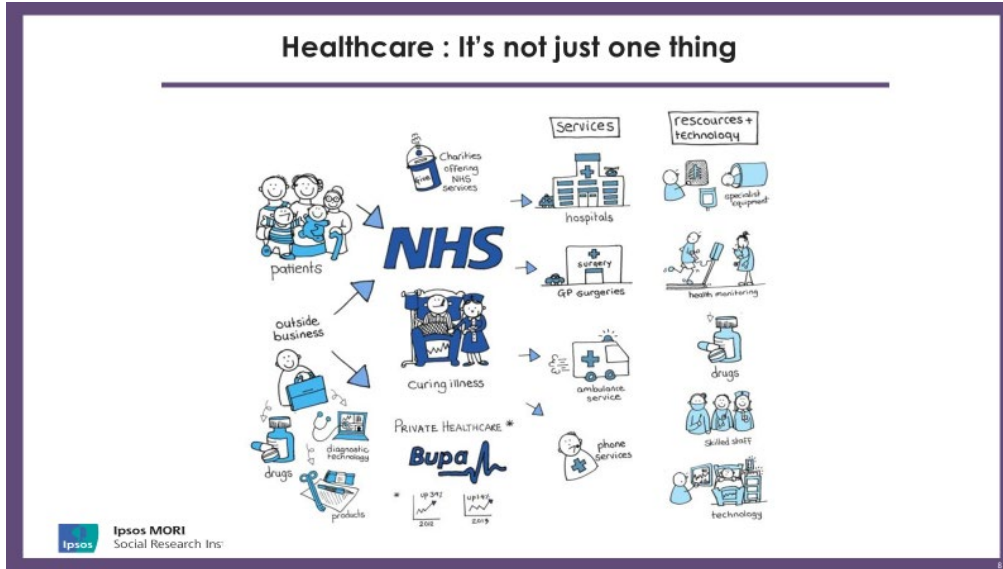


**Commercial users:**

- Community pharmacists
- Drug manufacturers
- App/imaging manufacturers
- Medical device manufacturers
- Insurance companies
- IT software companies



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### But what sort of data do they get?

Academic users and Commercial users

Different research studies need different kinds of data...

↓

CODED HEALTH INFORMATION

About diagnoses, tests and treatments, ethnic group and age

BUT no dates of birth, names or addresses

PATIENT ANONYMISED

Health Record for \_\_\_\_\_

NHS Number: \_\_\_\_\_ Hospital number: \_\_\_\_\_

Contact details	
First Name:	
Last Name:	
Address:	
Town:	Skipton
County:	Yorkshire
Post code:	SK72
Home tel. No.:	
Mobile:	
Further details	
Date of Birth:	11/79
Gender:	Male
Height:	1.8 m
Weight:	85 kilos
BM:	26
Smoker:	Yes

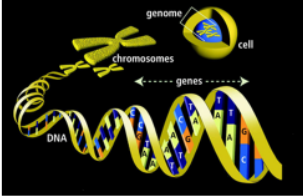
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# Genomics and the 100,000 Genomes Project



## Reading and interpreting a person's DNA (genomics) is revolutionising medicine


**1<sup>st</sup> sequenced genome: 1990-2003**





- **Scientists:** Thousands
- **Computers:** Hundreds
- **Costs:** £2 billion
- **Time:** 13 years

**2018...**

In the NHS, 1,000 genomes are sequenced per week

520 million messages 

6.9 million images 








22,000 episodes 

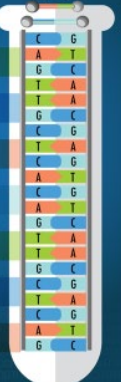
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### GENETIC TESTING

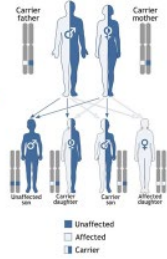

NHGRI FACT SHEETS genome.gov

**Genetic Tests Can Help to:**

-  Diagnose Your Disease
-  Pinpoint Genetic Factors That Caused Your Disease
-  Predict How Severe Your Disease Might Be
-  Choose the Best Medicine and Correct Dose
-  Discover Genetic Factors That Increase Your Disease Risk
-  Find Genetic Factors That Could Be Passed to Your Children
-  Screen Newborns for Certain Treatable Conditions



- Genomics says something about your **family, ancestry and descendants**
- Genes and the genome don't seal your fate

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## Genomics England helps deliver new scientific discovery and medical insights

100,000 whole genomes

➔

Better understanding of how disease works - **earlier diagnosis, and more effective treatments**

Anonymised genetic info

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## We are all genetically unique

## The more data in the pot the better

Patient	Sex	DOB	Race	Substance	Diagnosis	Genes in ClinVar	Shared Phenotypes
23300	4537	9.30	-0.5	De novo constitution	0.03	3	4 of 13: Microcephaly, Intellectual disability, Muscular hypoplasia, Hypoplasia of the tongue cartilage, Hydrocephalus, Frontal bossing, Microtia, Deafness, Brachycephaly, High anterior hairline, Microcephalic cranial defect, Oligosacchariduria and oligosacchariduria, Short stature
4663	4764	4.28	-1	De novo constitution	4.35	35	3 of 6: Intellectual disability, Muscular hypoplasia, Feeding difficulties in infancy, Oligosacchariduria and oligosacchariduria, Oligosacchariduria, No teeth
23308	4634	3.93	-1	Unknown	3.84	13	3 of 11: Microcephaly, Intellectual disability, Feeding difficulties in infancy, Hypotonia, Hearing impairment, Dysphagia, Psychomotor delay of the respiratory system, Broad forehead, With intervertebral discitis, Defect in the atrial septum
1851	4658	6.86	-0.9	De novo constitution	4.27	14	2 of 2: Intellectual disability, Polyphagia of the gastric contents
4665	4537	9.39	-1	Unknown	1.44	9	3 of 6: Intellectual disability, Feeding difficulties in infancy, Prominent nose bridge, Impaired hearing, Specificity, Deafness, High anterior hairline, Forehead ear hair, Hallow valleys, Asymmetry of the fingers, Long hair
23315	4634	6.40	-1	Unknown	0.11	1	1 of 6: Intellectual disability, Hypotonia, Macrodonia, Clonality of the 5th finger, Open mouth, Brachycephaly, Small pituitary, Autism, Asymmetry of the nose ear
27376	4634	6.32	-1	Unknown	0.22	1	3 of 6: Intellectual disability, High anterior hairline, Triangular face, Small for gestational age, Frontal bossing, Proportional short stature, Peripheral arterial disease, Long eyelashes
27192	4664	12.03	-1	De novo constitution	6.43	40	1 of 6: Intellectual disability, Cortical boneless hypoplasia, Strongly colonic megal, Deafness, Autism, EEG abnormality, Oligosacchariduria and oligosacchariduria, Abnormality of the face
4662	4764	1.03	-1	De novo constitution	1.53	1	1 of 5: Intellectual disability, Autism, Delayed speech and language development, Large earlobe, Headache

### D Developmental Disorder Gene ARID1B

ARID1B is on the Developmental Disorders Genotype-Phenotype Database (DDG2P) because variants in the gene have been previously associated with specific developmental phenotypes. Below is a summary of information about the gene and characteristics of the relevant causal variant. Further information can be found in the [DDG2P table](#).

Status	Mode	Disease	Consequence
Confirmed DD Gene	Monoallelic	Mental retardation, autosomal dominant 12	Loss of function
Confirmed DD Gene	Monoallelic	Coffin Siris syndrome	Loss of function

# Making genomic medicine part of the NHS



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## Who will genomic medicine be made available to first? (1)

- NHS will sequence **5 million genomes** over the next five years
  - The 100K Genome Project has sequenced c.90,000 genomes since 2013
- From early 2019, sequencing will made available to:
  - **Seriously ill children** likely to have a rare genetic disorder or children with cancer
  - People with one of **21 rare conditions**
  - People with **specific types of cancer**



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### Data about your genome (2)

- **Genomic data will be stored in several databases**
  - Consent is needed
  - It is not mandatory - there is an opt-out for data used in research and planning
- **Access to the data will include:**
  - Researchers in the UK and around the world, including commercial (for profit) organisations
  - Clinicians leading trials of new medicines, to identify individuals who may benefit from enrolment in the trial
- **Access will require approval from independent experts**
- **Data security will be as tight as it can be, but nothing is 100% safe**

### Data about your genome and primary care (3)

- A whole genome sequence result will be **delivered in around 12 weeks**
  - Over time it will happen more quickly
- It will be sent to the **person's GP or the patient's hospital** (along with information about family members, if tested together)
- **They will get training** to ensure they can interpret the results and make the best clinical decisions for them
- **Genetic counsellors will help the patient** understand the genomic result and provide emotional support

## Annex B

## Genomics: time for a new social contract? Public dialogue to uncover views of mainstreaming genomic medicine in UK

*Event 2: day-long session – 10am-4pm. DISCUSSION FLOW*

Time	Discussion structure	Questions and materials
10-10.15	<p>Introduction Warm up</p> <p><i>plenary</i></p>	<p>Arrival and registration, H&amp;S briefing 10 -10.15 Revisit introductory slides plus add slide on key aim for the session. We have 3 aims today</p> <ul style="list-style-type: none"> <li>Focus on a wider use of genomic medicine, in the NHS and beyond: your views of its benefits, any concerns you have, and any safeguards you'd expect</li> <li>o Discuss what might happen in practice and what you think is acceptable and not acceptable in different situations</li> <li>o Finally, decide on <b>what patients and the public would ideally contribute; and what they should expect back from</b>, a genomic medicine service – clinicians, researchers and industry</li> </ul>
10.15-11.05	<p>Benefits and risks of genomics</p> <p><i>tables</i></p>	<p>Introductions on tables <i>On tables divided by age</i> Review of post-task at tables</p> <p>Experts brief introduction (10 min) 3 mins each on their table. Participants question experts, assisted by facilitator. to be confirmed when we know who is coming: will share questions in advance</p> <ul style="list-style-type: none"> <li>• Experts explain their experience of / role in genomics / area of academic, clinical or business interest</li> <li>• What benefits do they see from a wider use of genomics in the NHS? What challenges will there be?</li> <li>• <b>What might change about our expectations of or experience of medicine? [issues relating to social contract]</b></li> <li>• What specific ethical issues do they see that they would like the public to talk about today?</li> <li>• For the public: what further questions would they like today to answer?</li> <li>• After last expert, each table feeds back to facilitator: <ul style="list-style-type: none"> <li>o Learned anything new/surprising?</li> </ul> </li> </ul>



Have your opinions on anything changed, or are you now thinking about any new questions?

**Spontaneous views on expectations, benefits and risk of genomics (30 min)**

- Spontaneously, what do you think will be the **top 3 benefits of this new medicine**? To you, your family and children, and then to society as a whole? Why? *Facilitator to capture on cards –one for each benefit*
- What are your expectations or assumptions as to what could be done with your whole genome sequence? What kinds of things do you think could be found out?

**Initial probe on issues around social contract** *Capture on flip chart*

- To have these benefits – **very briefly, what would you expect the public will need to do, or agree to? How acceptable would you find it to give your genetic data as part of your care?**
- How about giving your genetic data as part of the overall research process? Does your opinion depend on whether you would receive care, or not?
- Some scientists feel - **As a society we are at a tipping point - data must be donated and shared and we all need to be in this together to improve human health. A partnership is needed.** – what are your views?
- What **proportion of the population** do you think would need to share their genomic data to provide a really good service?
- Is this **partnership** different from the way healthcare has been delivered up to now? Will anything be different? i.e. how the NHS might behave, what you might need to do, what researchers or private companies involved in the system might need to do?
- What concerns do you have, at this stage? Are there any **unintended bad consequences** that might come from bringing a genomic medicine service into wider medical practice?

**(10 min)** Spokesperson from each table **presents back** their main benefits and risks and their views on expectations/ social contract.

11-  
12.20

**Exploring a case study family - to demonstrate how social contract might work in practice and tease out**

**11am – 12pm Exploring the trees**

To tell the story of how some people might engage with genomic medicine, we have built three family trees. Each table will look at one of these. The first questions relate to things which might happen as genomic medicine is used. This helps us to talk about some of the practical and ethical issues – we've been given the ideas for this part by the experts we have spoken with.

The second questions are ideas we've come up with ourselves, to push the boundaries a bit.

**expectations around it.**

Stimulus material: facilitator will have a blank flip chart page to start with and they will add stick-on pictures of the family members and summaries of key issues.

**For each tree:**

At each point participants are asked to reflect on what is happening to the character. Facilitator will follow the prompts below but will need to cover the following issues: **NB – these are all questions which relate to participant expectations of the social contract**

- What should the NHS help these people with?
- What advice and guidance should they be given, and by who?
- How should people hear about genetic results (letter, meeting, etc)
- At what point should these people be given choices about their care and about the use of genomics in the process?
- What is acceptable or unacceptable about this situation?
- Do you see any tensions here between different ethical concerns?
- If different members of the group disagree – if this happened in real life, how should disagreements be resolved?

**First tree: The Miller Family**

- 1) **Tom and Lauren have a baby with learning disabilities. The baby, Evie, has a whole genome sequence.** The data is kept on at least two databases, the NHS Genomic Medicine Service database and the research database Decipher, used by the NHS.

What safeguards should the parents and baby expect to apply to that data? Tom and Lauren could refuse their baby's data being added to the research database. How do you feel about this?

Note: Tom and Lauren will be able to opt out of their data being in Decipher (and according to Mark B, they will be able to opt out of their data being in the research arm of the GMS database, from what I understand). However, they won't be able to opt out of their data being in the clinical arm of the GMC database - this is going to be the way that they actually get a diagnosis, although in reality at the moment, the best way to get a diagnosis is to be in the Decipher database as this focusses on developmental disorders and learning disabilities. But basically the message of what you are wanting to get across is there are two types of database - clinical and research. Clinical is mandatory, research is optional, but to get the best individual outcome it is best to be part of both, but what comes with this are different levels of governance. Particularly for rare disease this is VERY relevant. If a kid is the only one in the UK

with a rare developmental disorder you want to be able to compare their genomic variant with data from Iceland, Australia etc and that's not going to happen from the NHS database. Other databases will have to be used in the NHS

- 2) **Lauren discovers that she has a BRCA1 gene alteration predisposing her to breast cancer.** This was an 'additional looked for finding' (as it's on the list of tests that can be given) in the sequencing of the genome of the three of them. She would be told the information if she wanted to know. Should she be allowed not to know? She is not certain whether she will get breast cancer. She has a history of anxiety and worries a lot now about breast cancer. She could have genetic counselling to identify whether she could just wait until Evie was older and test for her own sake.
- 3) **Lauren has two sisters, Gemma and Briony. Gemma is Lauren's twin.** They have a 50% chance of having the same gene alteration. Lauren tells them of her diagnosis. Gemma knows she has the same mutation. Briony refuses to test. Later in life Briony gets breast cancer which is treated at a high cost by the NHS. Overall, her care has cost much more than if she knew about the issue earlier (even if she'd had a mastectomy).
- 4) **Or – Briony does have it done, she has a preventative mastectomy which causes her much distress.** But she never knows if she would have got breast cancer at all. Genetic information in this case is about a propensity to have something, could there be a harm to Briony knowing about her diagnosis rather than taking her chances?
- 5) **Lauren has another sister Hannah but they've not spoken for years.** Lauren does not want to tell her anything about it. Does Hannah have a right to know she might have this? Does Lauren have a right to privacy? Does it make any difference if she also has a daughter?
- 6) **Lauren's parents Des and Elaine tell Lauren's aunt** and the aunt, Diana, asks for whole genome sequencing for her daughter, Emma – she is very worried that her daughter might get breast cancer. NB she won't get this, as it's not recommended - should she be allowed to ask for this, if it is not something she is offered?
- 7) **Evie's data is de-personalised and shared** with a number of researchers and included in large, international linked databases. Would you have any concerns around this? Commercial companies e.g. pharma developing treatments for some of her conditions have access to her data, including companies based in the US and China.

And more boundary-pushing examples....

8) **Evie’s data is kept forever.** Does this matter? She hasn’t consented. How about if in the future, genome sequencing can be done on people not just with learning disabilities etc but less visible conditions (diabetes, asthma). It would likely still be illegal for employers to ask for genetic test results. It was pretty firm under the old Data Protection Act, and likely even harder to justify under the new DP Act 2018 / GDPR. Is this enough for us – would we trust that these protections will protect employees? If data is leaked, even if illegal, would you think employers would look at it? Is it worth the risk for you, given the other considerable benefits of genomics?

9) **Evie dies quite young. Lauren and Tom want her data removed from the research database.** They can’t have this; it has been de-personalised and it cannot be removed from all the projects in which it’s been used, though can be removed from future studies. Should they have been told about this upfront, more clearly? Will people ever take it in? NB facilitator to make it clear that removing data from published studies is not feasible – studies might have been shared and used, can’t keep changing one percentage point etc.

10) **Diana doesn’t get the sequencing for her daughter, but she does end up in a research project which sequences her genome. She also goes to a private company and uses a DNA test to identify her ancestors.** She posts the answers on social media. Research has shown that it’s easy to identify people if they do this – if you’re a European-descent individual you get a third cousin or closer match, which makes the group of people small enough to permit re-identification. A PPI company (not based in the UK but which has access to her social media data) links her by name to the genomic information. Diana suspects (doesn’t know, but suspects) that this company now has her personal phone and social media data, the health record about her, and her genomic data. She is very worried about this. Like everyone, she gets cold calls about PPI but wonders if it’s because her data has been leaked. The company says that all records are kept separate. Should Diana have had help to weigh up the risks of putting her genomic information in the public domain? What kind of help is feasible?

11) In future, **Briony and Gemma are asked to disclose** whether they have had genetic testing for insurance – Briony has a higher premium as she has had breast cancer. Gemma has a higher premium because of the family history. The insurer takes into account Briony’s risk reducing surgery. But is it fair that they have the higher premium?

### **Second Tree: The Cooper Family**

Paulette and Hubert have two sons, Benjamin and Frank.

- 1) **Benjamin has a rare condition and is offered WGS in the new rollout of genomics.** Paulette and Hubert are worried by this - they've been scared by the Windrush scandal, which has affected some of their friends. They are concerned about anyone taking their data and "putting it in a database". Should they have the right to refuse the genomic testing? How can they be reassured?
- 2) **Frank's son Chris also has a suspected rare condition. He is offered WGS.** The results take a long time to come back – just over 3 months. Frank is very concerned through this time. What support should Frank and Chris get?
- 3) **Chris and Frank are both diagnosed with a rare condition that has no cure.** Chris is in good health through his teens and his symptoms are not too bad but as he gets older he gets more depressed about his future. He engages in risky behaviours which make him more ill, seems angry and upset. Should he have been told about his prognosis?
- 4) **Paulette's sister, Caroline, has a son called Jacob. He is fit and healthy, a keen sportsman, and works in the police force.** Jacob is offered WGS due to his cousins' health. But he doesn't want his genome screened. He is worried about future uses of genomic data. "I trust my colleagues but you always hear about the odd bad apple in the police. How about if I'm at a crime scene, and I work for a bad boss who wants to fit me up for a crime? With my genetic data out there, I might be at risk". There are no plans for the police to access health data. (And in fact Jacob could be 'fitted up' with a hair or cigarette butt at the scene) How reasonable is Jacob's concern? What sort of protections would reassure you if you were in Jacob's shoes? How should we balance concerns like these, relating to the future, with the benefits of genomics- for Jacob and for all of us?
- 5) **Suddenly, one day while playing football in his work team, Jacob falls down with a sudden cardiac arrest. It turns out that sadly, he is a victim of sudden cardiac death.** He has not had genomic tests but after death a genomic test reveals a genetic alteration. Could he have been saved with a screening? At what cost to the NHS of screening everyone? Alternatively, Caroline decides she doesn't want his genome examined. Does she have the right to do this, because if the death was to do with the genetic component, it might have a bearing on other members of the family? Assuming the rest of the family hadn't been screened, a different situation - should the NHS tell the family? And screen them?

More extreme examples: these are very future focused.

- 6) **Some years after Hubert's screening, he finds out he has a "Polygenic risk score"** which is a mixture of risks for how likely he is to get cancer. He has a 'lucky' combination of lots of gene variants that individually have no effect. The results are usually given

along the lines that someone is 2 or 3x more likely (or less likely) to have cardiovascular disease. So he takes these results quite literally and thinks he won't get cancer. He's been trying to give up smoking but joyfully now he starts up again. Within a year, he gets lung cancer & dies. Would he have been better off not knowing this? How should we inform and protect people about the way that this information can give people a likelihood of something? A further point – at the moment, scores are more accurate for people of Caucasian / European descent and not other ethnicities... does this mean we should inform people differently about this?

### Third Tree: Ahmed family

Nazir and Atef Ahmed have one son, Sadiq. He was married to Azra Ahmed and they have one child, Basma. He divorces Azra and gets married to someone else, Stephanie Ahmed {nee Smith}.

- 1) **Sadiq and Steph want to have a baby. But it's not happening, so they go for fertility treatment.** At the fertility treatment they are offered genomic testing. Sadiq has mild cystic fibrosis. The genomic sequencing confirms this has affected his fertility and he can't father a child. In the counselling session, Sadiq realises that Basma can't have been his child. What should happen from hereon? Who should approach Azra, if anyone? Has Azra's right to privacy been infringed? What help should they all get?
- 2) **Atef was offered whole genome screening when Sadiq was under 16,** to be part of a research study tracking children from birth. (e.g. risk factors for children from BAME backgrounds in socially deprived areas). She decided not to tell Sadiq and he was not screened. Is that fair?
- 3) **Nazir goes to 23andme and finds out that he has a genetic predisposition to Alzheimer's.** He is angry and upset and goes into his GP, demanding some kind of treatment right away. The GP says there is nothing that can be done, and maybe he can just try and keep as healthy as possible. If Nazir doesn't heed this advice, does he bear any responsibility for his health in later years? How about if Nazir decides to live life to the full, takes up some new hobbies, travels to Pakistan to see family, a trip he's been putting off for years? He has a better quality of life, living in the knowledge of the diagnosis. Is it acceptable for companies (or the NHS) to deliver results from genomic tests where there is no clinical treatment available?

### More extreme examples

- 4) **Steph and Sadiq do eventually have a child (through donor sperm), called Jadon. He is born deaf.** As he gets older he becomes part of an activist group championing the 'right to exist' of deaf people. His group claims that genomic screening of embryos is



leading to more mothers deciding to abort embryos which show genetic issues such as genes associated with autism, deafness, learning disabilities and dwarfism. He wants this sort of screening to be made illegal. Does he have a point?

- 5) **Steph, later in her life, develops bowel cancer. Her cancer genome is screened.** Fortunately, there are a range of drugs available to treat this particular kind of cancer. Steph is offered a particular drug by her local Trust. She goes online and finds that there are other drugs which are thought to treat the cancer better. Her care team explain that genetically, she's best suited to the drug she is on. She does not believe them and thinks she's just on a cheaper drug. She travels to the USA to try and get a different drug. Is it reasonable of her to try and alter her care plan like this? How about if it is actually a cheaper drug, but which treats the cancer well but maybe not quite as well as the specific drug for her genome, which American doctors recommend, but which is not recommended by NICE due to cost-benefit analysis? Should the NHS be offering cancer genome screening if it cannot offer an entirely personalised treatment regime?

**Final example -should always be done last after all 3 families completed – by each facilitator?**

**Steph has another baby. At this point, the neonatal heel prick test has been replaced with automatic whole genome screening of the baby.** All the babies are now part of a lifetime of potential uses of their genomic data (revisit talking head from data privacy). Is this a world which is acceptable, scary? Are there safeguards you can think of that you would put in place? Is it inevitable that such a world will come to pass, if we roll out genomic medicine?

At the end of the session facilitator to **prep the team to respond to plenary feedback**

**12-12.20pm**

**Feedback from each table**

- Most important issues you discussed
- Three principles for how genomic medicine should be used in wider society, based on your conversations
- IF TIME: what was different about genomics medicine in these scenarios compared to the dilemmas in conventional medicine

**12.20- 1.00 LUNCH:** after lunch, lead facilitator to do a washing line: One end of the room is "genomics in 10 years time is wildly successful" – the other end "genomics has led to undesirable social consequences" – please position yourself on this line  
Facilitator divides afternoon groups based on this.

The afternoon session will be focused on the social contract – then we will look at insurance and some other questions around data privacy.

1-2.10  
pm

Drawing up the social contract (NB this will also be considered in detail at the Summit)

(15 mins) Plenary discussion of social contract

- Have you heard of the term 'social contract'? What does it mean to you? What does it mean in medicine and healthcare, particularly?
- We could describe it as **"The shared, reasonable assumptions and expectations that everyone in the system has, which guide how the system works"**
- Why is this important (if you think it is important)?
- Who decides on how the 'social contract' works?
- Who has a role to play in making sure it is held to – government, citizens, patients, researchers, companies, charities... who else?

(35 mins) In tables, each facilitator has a flip chart. This is a large template that the table will fill out together  
Lead facilitator to explain the template in plenary.

Over the next 3-5 years we are thinking about people's **expectations** of genomics and medicine. What does everyone in the system have to do, to make genomic medicine a success? What would be additional 'nice to have' things that different players can do – and what are your red lines – what can never be done? In this section we want you to think about the **reasonable expectations** you have about genomics – what would create **good practice**.

Each table will have 3 audiences to work with, out of the 6 we have here, then we'll feed back.

- Group 1 look at NHS, patients, researchers
- Group 2 look at NHS, patients, insurers
- Group 3 look at NHS, wider public, UK government

	Who?	Have to do	Can do – nice to have	No way! Must not do.
1	The NHS and clinicians	<i>Example: give everyone access to genomic testing if their symptoms suggest they need it</i>	<i>Example: offer genetic counselling</i>	<i>Example: operate a postcode lottery</i>

2	Patients in the genomic service			
3	The wider public			
4	Insurance companies			
5	Researchers – in industry or academia (facilitators to tease our differences)			
6	UK government			

Facilitators to prompt, making sure to pull out the following issues:

- Is it acceptable that people can refuse to contribute their data to health research whilst benefitting (i.e. in clinical care) from the use of other people's data? How should people who refuse to share their genomic data be treated by the NHS?
- If genomics only works if there's a large number of people contributing, how do we make sure this happens? How about if the number required is a smaller proportion?
- Do you see giving data as a 'payment' for the genomic service?
- Do you see genomics as a mainly research process or a mainly clinical process (sort of – for society, or for you? A common or private good?)
- How should the public be involved in decisions on this going forward?
- How should the benefits of genomics be distributed – how do we make it fair?
- What are the expectations of researchers – how quickly do they need to benefit patients, or do they at all? Does it make a difference if they are funded through charity, industry, or the state? *NB increasingly health research needs to be undertaken by multi-disciplinary teams – because, broadly speaking, we've discovered the easy stuff*) so it's common for academic researchers to work with scientists and researchers in the commercial sector.

- Economically – how should we make money from this, if at all? If university research comes up with intellectual property – how should they decide to use it? How should they balance commercial value with patient benefit? *NB according to Professor Tim Hubbard, academic researchers get data for free while commercial researchers already pay for this.*
- How should insurance companies balance commercial interests with a duty to society (if in fact you think they have this duty).
- What are your expectations of UK Government in all of this? How should the UK government fund genomic medicine, given current pressure on the NHS e.g. ageing population? How should the government be involved in decisions on this going forward e.g. safeguards, code of conduct for insurers, giving organisations outside healthcare permission to access the genomics database?

**(20 min) Plenary feedback.** Each table feeds back their chart

- We have established something of a ‘social contract’. Do we need to write this down – or can we trust the NHS to run according to these principles anyway?
- What might help/prevent the NHS adhering to what we want?
- Who has the authority in the social contract as you see it? If the NHS has the ultimate authority to make decisions on healthcare, what does it need from the public to enable it to do this? And from other actors.
- Preventative future vs treating you when you’re ill - How far should the state secure our health, treat us, or help prevent us getting ill? Is it the job of the healthcare system to help us all to flourish – or to fix problems? Or something in between?
- Is a preventative NHS based on a different ‘social contract’ than the one we’ve had before?
- Do you see this social contract as more about your obligations and duties, or more about your rights and needs?

## 2.10-2.25 Break

2.25-2.55	Insurance	<p><b>Insurance</b></p> <p>New Code of Practice for insurers has just been issued – this is not a binding law but a code to which the industry signs up – it is nevertheless taken very seriously by the companies involved – the members of the Association of British Insurers which have signed up to the Code. NB if needed make it clear this is not about private medical insurance.</p> <p>Facilitator to hand out summary points on a handout</p> <ul style="list-style-type: none"> <li>• An insurer will not require or pressure an applicant to undertake a predictive or diagnostic genetic test in order to obtain insurance.</li> <li>• The results of a predictive genetic test may be considered in an application for insurance only when both of the following conditions are met:</li> </ul>
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1. This Code states that the specific predictive genetic test may be considered and;
2. The sum assured exceeds the financial limits set out in this Code.

### Going through the detail

- Before we all go through the detail, can anyone tell me what a predictive test is? How about a diagnostic test? *Facilitator asks expert to explain terms before moving on.*
- Is it right that you should not have to undertake these tests to get insurance? Pros and cons? If tests are there, shouldn't we use them to get the cheapest premiums for everyone in society? How about if you want to have your test considered? Should you be allowed to? What might be the wider consequences?
- NB – you can voluntarily disclose your test result if it's in your favour, for instance ruling out that you have a risk that's present in your family history.
- Is it important that predictive tests are considered differently from diagnostic tests? Why?

Facilitator shows second handout

Type of insurance	Financial limits above which predictive genetic tests may become relevant	Medical conditions for which insurers may ask for and take account of predictive test results, for policies above the financial limits
Life Insurance	£500,000 (per person)	Huntington's disease
Critical Illness Insurance	£300,000 (per person)	None
Income Protection Insurance	£30,000 per annum (per person)	None
All other types of insurance	Predictive genetic test results will not be asked for, or taken into account, whatever the level of cover.	

- These are the cases where insurers might ask for predictive tests – what do you think? Financial limits? Types of insurance? Types of conditions?
  - *Facilitator explain that Huntington's is unusual that it is a single gene, and there is a very high probability if you carry the faulty gene of 1) developing it 2) passing it to your children. Further information: there are around 4000 inherited disorders caused by single gene, or Mendelian, disorders. As noted Huntington's is one, others are cystic fibrosis and sickle cell anaemia.*
- Are there any conditions where you think predictive tests should be allowed? Are there any here you'd change?
- Any final concerns or questions about the use of genomic information by insurance companies?
- Further info on financial limits, if participants ask - 65% have less than £100K of life cover (probably because they are older term policies that decline as a mortgage is paid off). (will also bring along slide)

**NOTE FOR FACILITATOR:** We have the financial limits because any policies below this will never require disclosure of predictive test results. So we say that at least 95% of applicants can be 100% reassured.

Above the limits an applicant may need to disclose predictive test results if they are in the list of tests in Appendix 1.

Huntington's Disease for life insurance was the first application made in the old system (2001) and, because of the concerns this raised, was the only application under the old Moratorium.

It's possible that the ABI will make future applications for other predictive tests. These are likely to be single gene disorders, but may not be for life insurance. For example, critical illness policies pay out on a diagnosis of cancer so some of the familiar cancer risk genes are potentially relevant to very large CI policies.

We don't know when this might happen but the point of the annual report (and 3y review) is to be more transparent about levels of testing and industry views on the risk for insurance underwriting.

2.55-  
3.30

Different  
groups  
accessing your  
data – and

**Use of data about you; risks and privacy**

- Now we've thought a bit about how genomic medicine might be used, what are your expectations of the way data about you is protected, if you are taking part in genetic medicine?
- Who would be ultimately responsible for its safety?



## potential future uses of data

## Tables

- How do you feel about international uses? Commercial uses?

We have spoken to some data privacy campaigners and other commentators about the concerns they see; **here are two imaginary interviews** (NB, invented by Ipsos, a mashup of different interviews and perspectives)

### View from campaigners about some of the negative aspects of genomics (A)

Stimulus: Show talking head

- What surprised you – any food for thought here?
- Any concerns given this
- Any questions for experts
- How important is it that genomics helps us economically? How should the benefits of this be distributed? NB – new drugs and diagnostics are always developed by industry not in the NHS – could there be a proportional profit allocation? How should this work?
- What would be the ideal public engagement around genomics – now, and as it develops? This campaigner thinks that decisions about genomics could be a ‘slippery slope’ – that if we agree to one thing, then another, we might end up with something we don’t want (e.g. Eugenics). But shouldn’t we take each decision as it comes, and take into account how technology works and what happens, rather than deciding everything on principle? What do you think?
- If your data can be identified – does that change your views? Views are divided here – it’s likely to be a small risk and it would be illegal to re-identify data – how should the risk be calculated? Should the NHS prioritise keeping data private, or some of the other benefits of genomics? How should they decide?
- If industry is needed to make genomics work – how do you feel? Is it necessarily a good/bad thing? *Facilitator to give example of positive work between industry and genomics - Alexion, a company validating a target gene for the development of new drugs for a rare childhood kidney disease. Usually people with this disease die before they are 15. Alexion have identified 10 undiagnosed patients in the GE Rare Diseases Cohort who carry the disease. They have given the information back to the Genomic Medicine Centres the children can now be treated with current best practice - hopefully preventing or delaying the onset of kidney failure. It also allows for genetic counselling on any risk to future pregnancies.*

### View from data privacy campaigns (B)

Stimulus: Show talking head

- What surprised you – any food for thought here?
- Any concerns given this

		<ul style="list-style-type: none"> <li>• Any questions for experts</li> <li>• If you were part of the research pool what would be your expectations – of knowing about findings, of having treatment if necessary?</li> <li>• How do you feel about a national DNA database? The point is not that the databased is designed to be used by others outside the health sector, but that it might be in future, once constructed.</li> <li>• If data is lost or leaked – what might be the harms to people?</li> </ul> <p><i>Facilitator to capture thoughts on flip charts</i></p>
3.30-3.40	<b>Final post it exercise / green dots</b>	<p>Looking at all the flip charts we created earlier, expressing our expectations for a social contract; go and take a look and add a green dot to anything you think is particularly important.</p> <p>Also, add a post it note if anything has been missed – in the light of the discussions we just had about privacy, potential future uses of data, insurance etc.</p>
3.40-4.00	<b>Wind up / housekeeping</b>	<ul style="list-style-type: none"> <li>• Housekeeping re. final Summit</li> <li>• Event questionnaires</li> </ul>

*Facilitator notes on some benefits / probes*

*Individual*

- more precise, personalised healthcare [ability to identify what drugs etc work on different people]
- tailored treatments [ability to identify e.g. how cancers respond]
- improving life for people with long term conditions [future inclusion of things like asthma in genomic research priorities]
- cures for diseases we can't currently treat [e.g. maybe heart disease, asthma, diabetes, irritable bowel, autoimmune disorders?]
- better diagnoses if we can't treat something [e.g. the Charlie Gard type cases where there isn't consent on the diagnoses, that might become rarer]
- getting your medication right [stopping you taking something that you might have taken for ages but never worked as well as it could have done]
- speeding up medical science [saving money and time in the NHS]

### Wider society

- Understanding more about the diversity of human behaviours [e.g. genetic causes of mental illnesses – could this help us be more tolerant as a society?]
  - Changing understanding of what the genome means, how the body works [could this lead us to live generally healthier and happier lives and make better choices for ourselves?]
  - Chance to carry on world-leading centralised medical system to the next stage [UK is in a good position to do this given the way the NHS is funded and run? Emphasise the point that industry and NHS can work together successfully]
  - Targeted service planning [knowing more about the population gives us the ability to understand future needs of population?]
  - Understand the link between disease, genes and environment across the world [make sure to say that to do this, we need to access VERY large datasets of linked genomic and health data - not just within the NHS, but globally, between countries and across geographical boundaries]
- 
- Are there any other benefits you see – maybe other ways a set of data about our genomes could be used? *NB prompt gently on e.g. police usage of a database collected for health use, as we do not want to get into dystopian future uses – but do want to capture any sense of unintended consequences that the public would want to be further consulted on*

Page 1

Genomics is already a done deal. The government wants to make money from biosciences, one of the industries where we can be world leaders – it's part of the wider industrial strategy.



This means that commercial companies are working together with the NHS really closely. Once the infrastructures are built and we depend on the companies to deliver our services, will we be able to change our minds?

And the public haven't been involved in the discussion – recently, the government announced the future sequencing of 5 million genomes without public consultation.

It's not really possible to de-identify genomic information – it's usually possible to re-identify the individual. So that's something we should be told about and something the public should discuss.

Page 2



Rolling out genomics into the NHS will result in a population-scale, centralised, biometric database. It'll be great for genomic medicine but it can also be used for all sorts of other things. We need to make sure we know the risks.

When you create a big database of tissue samples and sequenced DNA you create a database of the population which is instantly available for other identification purposes.

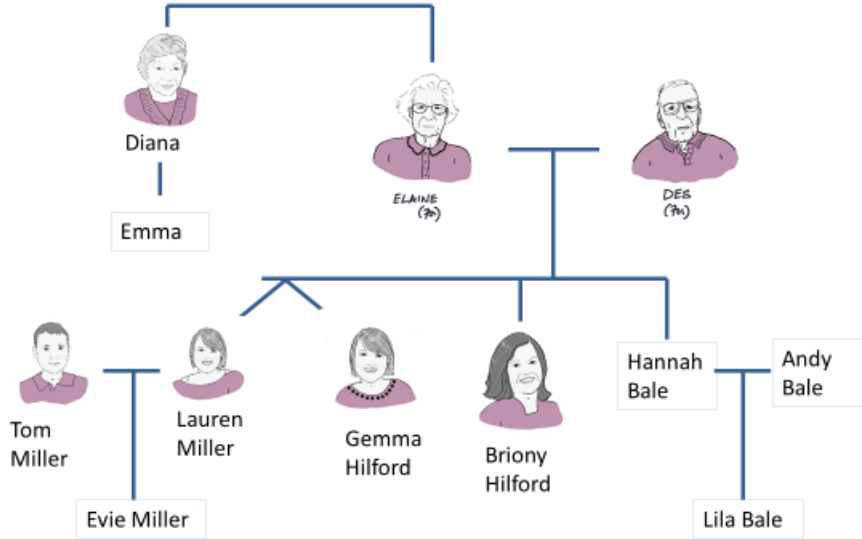
People will always say "we won't let anyone use this outside healthcare" – but if politicians, for example, decide that the Home Office or Police can have access, we can't do anything about it.

The cost of genomics is still going down and down. Why invest in something that might be outdated soon, and provides this dangerous grouping together of data?

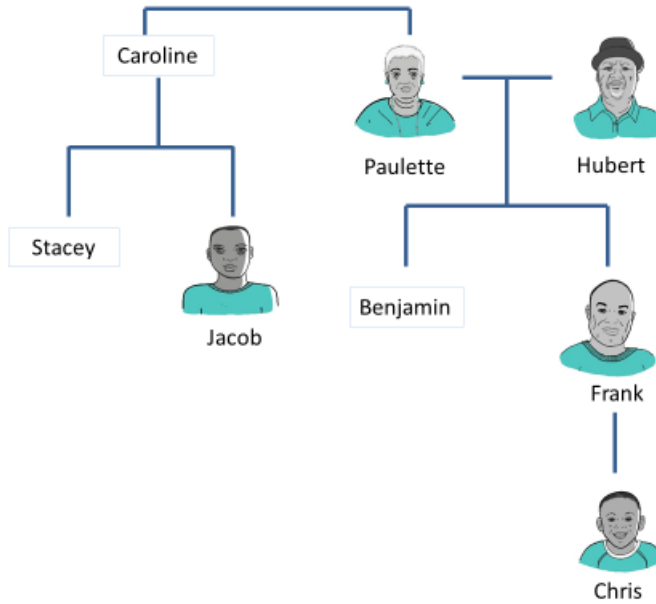
Let's wait 5 years and improve the NHS so it can really deliver a 21<sup>st</sup> century, person-centred, digitally-based level of care.

I am concerned that if we create this database and then people are scared about whether their data will be safe, people might not actually go to the doctor when they are sick.

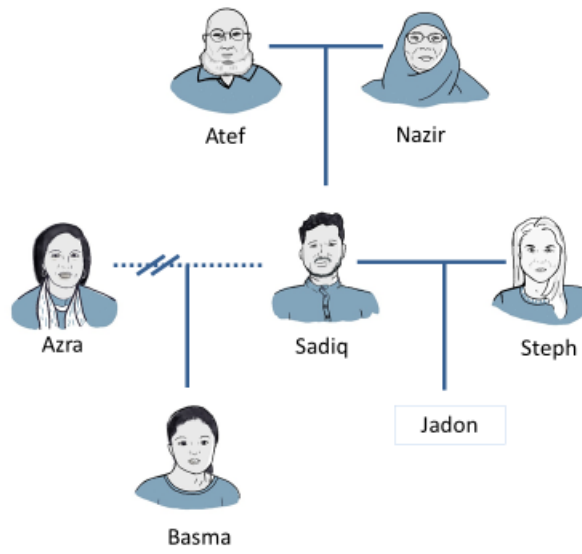
### Miller Family



### Cooper Family



## Ahmed Family





## Annex C

## Summit structure

- c.21 public participants, c.21 stakeholder/OG/patient panel voices
- Refreshments/biscuits etc on arrival

Timing	Groups	Area for discussion
12-12.30pm	Plenary Then groups split by stakeholder type to warm up so public participants aren't intimidated. Possibly 4 groups only at this stage.	<p><b>Presentation:</b> what we've learned so far: few thoughts from all the locations. Presentation from Viv Parry / Ann Dalton reminding us of key themes, why genomics is important/different from other forms of healthcare. Show film.</p> <p>Today we are talking about expectations around healthcare in the future of genomic medicine, and the values and principles we all want to preserve. Emphasis on collaboration and ensuring that everyone's voices are heard.</p> <p><b>Exercise:</b> picture sort [lots of abstract and non-leading images] – picture that makes you think about the values of healthcare, or the shared social understanding we have. Picture of hope for future, and concern for future. – probe on how genomics fits into that.</p>
12.30-1pm	Tables split into new groups - mixture of ppts from across locations, and stakeholders	<p><b>Introductions of new tables.</b></p> <p><b>The values on which a social contract is based and the founding principles of the NHS</b></p> <p>What would you say are the founding principles of the NHS? What values are important to the NHS and us, as citizens? (facilitator to collect)</p> <p>Then we present the founding principles (as in Generation Genome)</p> <p><b>1) availability of healthcare on basis of need and independent of ability to pay</b></p> <p><b>2) healthcare of a high standard of excellence</b></p> <p>Spontaneous response: What are your thoughts on these principles? How do these fit with today's 21st century world of healthcare? Is there anything else happening today (outside genomics) which affects these principles? How should we protect or amend these principles, to accommodate genomic medicine?</p>

		<p>How does these principles relate to you in a world of genomics – what are the rights you feel you have, and the expectations you feel are placed on you?</p> <p>What would you say the rights and expectations and obligations of the public in general are, to make this work today? Of the government? Of the NHS itself?</p> <p>What principles do we want most to preserve in healthcare of the future/world of genomics? Each group collect <b>values/principles for future healthcare/</b> genomics&amp; we cluster on a wall/flipcharts.</p> <p>Plenary feedback</p>
1.00 – 1.35-pm	Lunch break	& do vox pops
1-35. 1.45pm	Plenary – lead facilitator presents next exercise	<p>Around the room we have <b>5 stations</b>. They relate to different areas where genomics could make a difference to our healthcare. In each area Ipsos MORI has invented ways to bring to life some of the ideas we discussed at the previous stages of this dialogue.</p> <p>These are all made up by us – they're not future policies or plans from Genomics England – just ways to talk about positive and negative things that could happen if genomics is used in different ways.</p> <p>We're going to discuss what we think of these ideas and also consider all the players in the system – patients, the public, government, researchers, the NHS, clinicians...</p>
1.45- 2.40pm	Each group rotates around first 3 stations	<p>Our 5 facilitators stay by their stations, while <b>each group (made up of public and stakeholder participants) rotates.</b></p> <p>1: 1.45-2.05pm</p> <p>2: 2.05 – 2.25pm</p> <p>3: 2.25-2.40pm</p>
2.40 – 2.50pm	Short break, get drinks, continue	
2.50- 3.30pm	Each group rotates around the next 2 stations	<p>4: 2.50-3.10pm</p> <p>5: 3.10- 3.30pm</p>

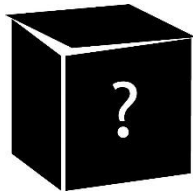
3.30-3.50pm	Return to sit down in table groups	Summary of most important principles the group thinks should guide development of genomics in the NHS. Facilitator prompts on all areas & particularly focuses in on any issues arising from their own station. Participants use their own booklet to feed back their thoughts.
3.50-4.00pm	Plenary	<p>Present back – each facilitator/group presents back on their area.</p> <p>Final evaluation questionnaires completed (including evaluation of whether and how participants feel their views have changed – both public and stakeholders).</p> <p>Wrap up – senior GE speaker to explain what happens next, thank participants, and share one or two thoughts on what they will personally take away from the day.</p>

## The five stations

NB: stimulus for this is intended to create metaphors to start discussions – we'll be really clear that it's Ipsos who have invented this. We really need something tangible to help people think about the abstract issues which underlie potential changes to the social contract.

Each of these areas brings to life a **tension which has been identified at previous stages of the project** – where participant expectations clash with what genomics must include, or where the exact way genomics should be rolled out needs to be bottomed out further.

At each station the facilitator demonstrates our stimulus material and asks questions.



### Station #1: Where is our data?

Ipsos facilitator: Graham

Experts: Natalie Banner, Understanding Patient Data; Phil Booth, MedConfidential

**Uncertainty:** What are your thoughts on the uncertainty of not being able to entirely pin down exactly which 'genomics libraries' our data might be part of, in future?

- **Stimulus:** *A black box* with icon of your GP/hospital on the outside. Inside there are pictures/cards/things representing China, America, Pharma companies, researchers from different places. Examples of health record and digital versions of genomic data files. Also include examples of face id on smart phones, posting on social media.
- **Discussion:**
  - What is your data? Looking at the non-health data in the box. Thinking about data generally, how far do you know where data ends up? (e.g. your phone data, social media) How much of a concern, if any, is this to you? How does it affect your behaviour?
  - You give your data when you consent to genomic test at GP/hospital. What actually is health data about you? Looking at different examples of data, health record, file of genomic data. Is this how you imagine genomic data to be? How do you imagine genomic data looks, how is it stored? Make the point that you need skills to reinterpret it. Thinking about this kind of data, where do you think it might end up?
  - At previous stages in this project people have said they're only happy with giving their data if it's never "taken off the NHS servers" – well, GE is a kind of commercial company working alongside the NHS so it can be hard to generalise about 'commercial access'; pharma companies and others are involved (as per stimulus) and there are other places data can end up – internationally, plus there may be a need to share with other private enterprises to develop genomic research or clinical treatments. What kinds of "genomics libraries" are we imagining? What, if anything, would have to be in place for you to feel comfortable that your data is in this place?

- What are the rules and expectations for how everyone should behave around data? What conditions do you think people/organisations should have to comply with to show they are safe custodians or users of data about you?

**Reidentification and risk:** Pushing participants further on the idea that reidentification is always going to be a possibility – are they really aware of the tradeoffs, and how do they think society should weigh these up?

- *Stimulus: Summary/picture/poster of Yaniv Erlich paper on how easy it is to reidentify*
- *Discussion:* Data security: Yes, it is illegal to reidentify data. But it's not as simple as saying "I will only give my data if it can never be reidentified". This kind of data will always be possible to reidentify with enough bioinformatics skill. how happy are we to live with the risk that reidentification might be possible? Do you need total certainty it won't? How would you describe the level of risk? Who should assess that level?
- What would reassure you that the chance is minimised? What governance processes do you think should be in place to deal with any breaches? How important is the 'punishment' for breaching data security for you?
- *Possible input from Edward Hocking/ MedConfidential?*

**Summary for this station; on what the 'social contract' should include:** Given that we can't guarantee entire data security, what should we expect from patients, public, NHS, government in the new genomics rollout? How well can we adhere to the principles and values we came up with earlier? What kinds of safeguards around data can we expect?

**Comms:** How much do we need to know about data security? Who's responsible for informing us?



## Station #2: The patient of the future

Ipsos facilitator: Emily

Experts: Jonathan Montgomery, University College London; Jayne Spink, Genetic Alliance

**Personalised and predictive medicine:** How will personalised medicine change the way we need to act as patients, clinicians, members of society?

- **Stimulus: *Cartoon of future patient***, labelled with features of the future patient. These are knowing their polygenic risk scores for risk of diabetes, therefore being monitored. Having access to information about their children's risk of diseases as well. Having access to professionals to give them advice on how to weigh up the risks and benefits of learning more about their genome. They take a drug on prescription, their doctor gives them one rather than another because they know it works better for that particular patient.
- **Discussion:** How will knowing more about our own profile change the way we behave as patients?
- Will we have greater expectations of the NHS, greater expectations of our own personal responsibility? How should the NHS system, government, clinicians, help patients in this world
- What expectations do we have about the professionals who will help and advise us on learning about our genome? We've heard about genetic counsellors – there are only around 200 of these in the health service. How do we imagine the clinician of the future will be able to help us?
- How do we prevent a dystopian reality where people are 'blamed' for having 'bad genes' or for not taking action to improve their own health?
- How about uncertainty – a 'learning healthcare service' won't necessarily be able to tell us exact risks for this or that, and genetic knowledge might lead to more complexity. What should we be told about this? What happens if the media are always telling us about certainty – "A gene for x" – is often much more deterministic than there is evidence for. How might this affect our perceptions of what's possible in the system? How should we describe and deal with uncertainty?

### Free riders

- **Stimulus: *free riders***: cartoon of someone hitching a ride on a bus or similar – we explain the metaphor of a free rider.
- **Discussion:** how does the system manage 'free riders'? What's fair? How do we communicate the idea of a 'tipping point' in data where everyone needs to get involved?

**Dystopian futures:** Where are the limits of how we should use genetic information about ourselves?



- **Stimulus:** Mock up headline “Dubai sequences genome of all city’s residents to prevent and mitigate diseases” and “Scientists can edit embryo to resist HIV infection”
- **Discussion:** What rights should the patient of the future expect to have about making decisions based on the genetic information we know about ourselves? Are there some things we should not be allowed to know or to act on? What do we think about risks such as making decisions which result in losing whole groups of people - those with Downs or deaf people for example? Is this a form of eugenics? Who should stop this happening?
- In the Dubai example, it could potentially be mandatory to have your genome sequenced; in future it would be possible to segment or divide the population based on their genome. What do we think about population-wide screening, could it be used e.g. to deny healthcare to certain groups?
- In the Chinese example, the scientist has edited a gene he claims would lead to greater protection against HIV. The babies have just been born but their health prognosis over the long term obviously isn’t known yet. How should we start to use this information in clinical practice? If we don’t know what might be the knock-on effect of editing one gene? Or do we have the right NOT to do it, if we know we could, for example, edit out sickle cell anaemia genes?

**Summary on social contract:** What changes about the roles and responsibility of all the players in the system?

**Comms:** How far do we, as members of the public, need to know explicitly about any changes, or can we just infer them or experience them? How far do we need a shared mandate for our roles and responsibilities in this changing world’?



### Station #3 One big family?

Does our understanding of what it means to be a patient change?

Ipsos facilitator: Sam

Experts: Ann Dalton, Sheffield Children's Hospital; Fiona Crowe, Genomics England

- **Stimulus:** The patient is the whole family -picture of a big family: <https://www.theguardian.com/science/2018/nov/25/woman-inherited-fatal-illness-sue-doctors-groundbreaking-case-huntingtons> Article in the observer headline/summary, plus quote from the Huntington's case lawyers – "Should clinicians be legally obliged to consider the interests of anyone they are reasonably aware of who could be affected by genetic information – or is the protection afforded by current professional guidance enough?" also quote from the journalist "In future, a patient may be not just the person who provided a genetic sample, but may be defined as also those affected by that genetic sample"
- **Discussion:** current GMC guidance says that one reason to disclose relevant information to others without consent is to "prevent serious harm in others" (current GMC guidance).

*Facilitator note: do give quick summary if think would help ppts grasp the issues: Guidance for doctors on disclosing personal information (adapted from General Medical Council guidance)*

*Confidentiality is an important ethical and legal duty. Disclosing personal information breaches a doctor's duty of confidentiality unless the following applies:*

- The patient consents, implicitly or explicitly, for the sake of their own care or for local clinical audit*
  - The patient has given their explicit consent to disclosure for other purposes*
  - The disclosure is of overall benefit to a patient who lacks the capacity to consent*
  - The disclosure is required by law or the disclosure is permitted or has been approved under a statutory process that sets aside the common law duty of confidentiality*
  - The disclosure can be justified in the public interest*
- What kind of serious harm would you imagine is relevant here – who should decide?

- Would you expect with genomics for familial information to be shared - by whom, and with whom? Should clinicians be saying “familial risk” rather than disclosing individual info? How should we separate personal clinical information from the familial information that led to it? What should patients be allowed to decide about the way their information is shared? Should a patient be able to decide not to let the family know? Does that put the doctor, a difficult position? How about the researcher, if we are in a situation where the researcher might have a clinical duty to feedback findings which are relevant to care? How do we do ‘the best healthcare we can’ under these circumstances?

### What support do we get?

- **Stimulus:** comfy chair, genetic counselling leaflets?
- **Discussion:** What should be the role of genetic counsellors and others involved in giving genetic information to patients? How important are they in the system? How should counsellors work with GPs and other healthcare professionals? What happens if people can’t understand what they are ‘consenting’ to? How should we protect vulnerable people who give their data and take part in genomics – what systems do we need in place for this? If someone’s harmed, what redress do we expect?

**Summary on social contract:** What changes about the roles of a patient and a member of society, in the new world of genomics?

**Comms:** How far do we, as members of the public, need to know explicitly about any changes, or can we just infer them or experience them? How far do we need a shared mandate for any changes to how we understand who is the patient?



## Station #4 Show me the money

Ipsos facilitator: Polly Experts: Rose Grey, Cancer UK; Monika Preuss, DHSC

**How can we build trust in the NHS' ability to deliver this service, in a time of austerity?**

*Stimulus:* Matt Hancock's announcement on sequencing 5 million genomes

*Discussion:* How should this play out in terms of the genomic counselling needed; how would we deliver this fairly? What role should individual Trusts have in the rollout?

*Stimulus:* The NHS – it's not just one thing! Plus, logos of pharma companies

*Discussion:* Is it important that the public sees benefits in their care, at the same rate as we see advances in research? Is it 'fair' for us to give our data if the balance of investment is in research and patients do not see the fruits of this for a long time? Do you see this as something that benefits our research capacity, or our clinical capacity? Who should make money from genomics? Is it fair to expect private companies to invest and see no return? How should government, society and others value the data which genomics relies upon? Can we expect government to invest enough money on its own, to secure the benefits of genomic medicine?

### **Private sector genomics**

*Stimulus:* 23and me leaflet offering Black Friday deals

*Discussion:* What should be the role of data we collect ourselves, or share ourselves, in the system of genomics? Whose responsibility is it to protect us from risk? (e.g. giving all our data to online service where it's not as secure or could be repurposed in future).

**Summary on social contract:** How should we pay for genomics?

**Comms:** What do we need to know about the way the genomics service is funded? What would we need to be assured of to be happy with the NHS' ability to deliver?

## Station #5 Blurred Lines: research and clinical practice come together in genomics

Ipsos facilitator: Sarah

Experts: Michael Dunn, Wellcome Trust; Pauline Mullin, Medical Research Council



## Different roles for clinicians/researchers?

- *Stimulus:* a pretend/mock up blood test and consent form and/or two pictures, a researcher and a clinician, with a dividing line between them or an open door...a white coat someone can put on and a stethoscope so people can pretend to be researcher /clinician... Then a sheet of cardboard with holes in – representing a blinkered version where only some information is available to the clinician and where the researcher has limited options to help the participant. We will explain how this works!
- *Discussion:* what does 'being a good researcher/clinician/patient' look like in this new world? What does 'respect and care' look like?
- At the moment, (very crudely) the researcher doesn't necessarily have to think about your care and the clinician doesn't have to think about the research. They operate under different rules, for instance research requires HRA/REC approval and clinical practice doesn't. Even if the patients don't mind whether they are dealing with a researcher or clinician, the researcher or clinician may feel they have to be in one camp or another, because of the governance arrangements for each.
- This is important for patients because researchers and clinicians having different roles affects the way data about you might be used and shared. Ideally we might want our data to be as tightly controlled as it is in one research study – or only accessed by our clinician for our care. But this new science needs our individual records to be linked up and for the old permissions for researchers / doctors to share information to change. Are we happy with the trade off, or what reassurance would we need? Would we prefer the blinkered vision where data is shared less and we use the 'old rules' for clinicians and researchers? We need to compare the limited roles with the tighter data restrictions with the more open data sharing and greater access from both sides.
- Genomics can yield information which helps public health, by comparing individual data with other data, and can help individuals, if knowledge gained from a whole dataset is applied to one individual. What will change about the researcher's role if something is discovered which impacts the care of the research participant? How should clinicians keep abreast of what is happening in research?
- Some academics have said that being a research participant in future won't be about consenting once, e.g. when you have your blood taken. It'll be more like 'entering a relationship' where things about you are known, and into the future more things may be learned. How do you feel about that? On consent, at previous events, you said "Don't share data in ways which are not consented" – but we also recognised we can't always consent to everything before it happens – in the case of the research discovering new information, how should researcher and clinician behave? What would be a reasonable and valid level of consent, if it's not possible to identify all the ways the data might be used in future? To be valid it needs to be informed consent, voluntarily given.

### Different roles for individuals in society

- **Stimulus:** something representing a crowd. Picture of *Field for the British Isles* or similar?
- **Discussion:** How much do you need to know about the fact that research – helping other patients – might need access to your individual level data? How do you feel if you don't personally benefit from this? What should you expect from doctors, researchers, as a 'data donor'? Does this change if the study is done by a private company?

**Summary on social contract:** What changes about the roles of a researcher and a clinician? How important is that change to you, the public – and what do you want to hear about it?

**Comms:** What messages do we need to hear about the role of the individual, each of us, and society, all of us, in genomics? Who should be telling us about this?

### Individual feedback: stimulus material

Public participants are also given a small booklet – **My thoughts about genomics** – to fill in as they rotate around these stations. We reserve a few minutes to do this each time we rotate. Stakeholders can also have this, to record their reflections / any surprises at each station. We collect them in at the end. Contents are as follows:

- Page 1 Where is our data: Genomics cannot guarantee a completely risk free sharing of data- because for it to work, data must be shared. What would need to be in place for you to accept the risk of reidentification from your data?
- Page 2 The patient of the future: What is fair when it comes to contributing data: should people be allowed to benefit from genomics if not contributing data themselves?
- Page 3 One big family: If, in the future, we have to think about the patient's family as well as the patient, how does this change our relationship with our clinicians, and what we expect from healthcare? How important is it that the public as a whole knows that how we think about 'being a patient' might change?
- Page 4 Show me the money: How should we pay for genomics? What do you need to know about the way genomics is funded, to be reassured that the service is running well and fairly? How far should the government be investing in genomics, and how far should others?
- Page 5 Blurred Lines: What changes about the role of a researcher, and the role of a clinician? How important is it that the public knows about these changes?



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