

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



Rapid Literature Review

Graham Bukowski, Sarah Castell, Emily Wayne, Samuel Yetman

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Summary of findings

Background

Genomics England with support from Sciencewise, has 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 involves exploring the principles the public believe should underpin any 'social contract' between the public and providers of genomic medicine.

The dialogue is intended to build on existing research in this field, and fill gaps in public attitudes work which has already taken place. A brief and rapid literature review has shown us what is already known by the public, and also the views of experts, clinicians, and patients on some of the 'live issues' in genomics.

The documents we assessed highlight areas where further public debate is needed. In this document we identify **seven areas** where the literature gives us a head start on which issues should be covered, what is known about them already, and the implications for our dialogue.

Key implications of what is already known, for this dialogue

1) Awareness of genomics is low; we should ensure that fundamental concepts around data, science, research, and the healthcare system today are understood; discuss what genomics can and cannot achieve, and what will happen if public support is absent; and use Understanding Patient Data (UPD) terminology for clearest expression of concepts.

2) Younger people and ethnic minorities have different concerns around genomics from other audiences; these should be accounted for. We should, potentially, sample for young people, and reflect BAME audiences in the sample as a whole, plus ensure our materials give specific case study examples relating to the concerns of BAME groups.

3) The public perceive a range of individual, also longer term societal, **benefits and risks.** We need to present examples of how these might play out to work out how they **assess and balance** them.

In particular, there are **uncertainties** specific to genomic research and treatment, where risks and benefits are not certain. These include the **scope** of genomics' impact, the possibility of diagnosis without **certainty** of treatment, the **speed** of uptake of genomics, and longer term social changes to the **fabric of society** (for better or worse). The current dialogue can explore these more systematically than previous research has achieved.

4) There is a need to deepen our understanding of where public 'red lines' are on the following issues:

- How **incidental findings** should be handled; we could explore legal issues, timing of information, plus ethical issues around impacts on the family (ancestry, finding out non-

genomic information, information about babies and young people, different cultural understandings of kinship).

- Issues around the processes of **consent**: how **reasonable consent** can be achieved given the uncertainties of future data use, the ethics of automatic opt-in, questions of equity around dynamic consent, and the role of advice and counselling.

5) Issues around **access to, and use of, genomic data**. Previous research has looked at commercial access to health data from a number of different angles. This study should focus on issues specific to genomics, such as:

- particular risks to individuals and society around **insurance** uses of genomic information
- the role of **public and private** partnerships in delivery of genomics
- **international** data sharing, when the data is genomic data

...and exploring these issues in the light of different **age and socioeconomic groups**.

6) Public, patients and clinicians are all concerned about the **current and future capabilities of the NHS**. The success of genomics will be determined by how it can be deployed within the constraints of the existing healthcare system. The dialogue could discuss public expectations around: how **hybrid** models of research and care could work; how the NHS workforce can be **upskilled** to meet demand; and how the NHS should demonstrate its **data protection** capabilities.

7) **Communications, language, and terminology**: there is a good background knowledge of principles already, gained through a number of studies and reviews. This dialogue should focus on what the public need to know about a **national level rollout**; covering increasing awareness of the potential of genomics, its uncertainties and risks; the level and detail of information required, and the different needs of particular groups.

1. Background and Objectives

Genomics England with support from Sciencewise, has 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 involves exploring the principles the public believe should underpin any 'social contract' between the public and providers of genomic medicine. The dialogue is intended to build on existing research in this field, and fill gaps in public attitudes work which has already taken place. This rapid literature review therefore synthesises the findings of recent research, on attitudes to genomic data and related issues. It will be used to inform the design and scope of the public dialogue workshops, influencing such questions as how the project is sampled, the types of materials shown and the areas we cover in discussion.

This review has a small scope and tight focus. It identifies specific gaps in previous research scope, plus any insights which raise questions and need further exploration. We would note that this is not a comprehensive meta-analysis of all research done on attitudes to genomics.

1.1 Scope of review

Document selection

This review draws on literature in various forms, including policy briefings, qualitative interviews with the public and clinicians, public surveys, research summaries, legal briefings, and media and watchdog perceptions of data security.

The selection of documents prioritised studies relating to BAME participation in genomic medicine and broader medical participation, in recognition of their significantly lower uptake of and trust in these services.

Ipsos MORI, Genomics England, and the project's oversight group all contributed suggestions of relevant research. Iterative discussion reduced the list to ensure a reflective spread of literature across our main interest areas. We have also substantiated some comments with further hyperlinks relating the findings to other research undertaken by Ipsos MORI and others, to set the findings in an even broader context.

The review includes the following documents¹:

¹ Given the makeup of the oversight group we did not explicitly include *Generation Genome: the Annual Report of the Chief Medical Officer 2016* as one of our documents. However this has been a key document for shaping the overall thrust of the project. Its contents and conclusions continue to shape our thinking.

#	Author	Title and date	Topics covered
1	A Allford et al	'What hinders minority ethnic access to cancer genetics services and what may help' (2014)	BAME participation
2	P Chow-White et al	'Knowledge, attitudes, and values among physicians working with clinical genomics: a survey of medical oncologists' (2017)	NHS capacity
3	S Dheensa et al	'Towards a national genomics medicine service: the challenges facing clinical-research hybrid practices and the case of the 100 000 genomes project' (2018)	Ethics; Consent
4	Genetic Alliance UK	'Genome Sequencing: What do cancer patients think? Patient Charter' (2016)	Participation of patients with rare diseases
5	Genetic Alliance UK	'The Genomics Conversation: Survey Results' (2016)	Public attitudes
6	Genetic Alliance UK	'Genome Sequencing: What do patients think? Patient Charter' (2015)	Participation of patients with rare diseases
7	Genetic Alliance UK	'What do patients with rare genetic conditions think about whole genome sequencing in the NHS? Research findings for the 100,000 Genomes Project' (2014)	Participation of patients with rare diseases
8	GfK NOP	'Ethical Issues Relating to Involvement of Cancer Patients in the 100,000 Genomes Project: Qualitative research findings' (2014)	Ethics; Participation of patients with rare diseases
9	A Middleton	'Society and Personal Genome Data' (2018)	Social contract
10	Academy of Medical Sciences	'Exploring a new social contract for medical innovation' (2015)	Social contact
11	Ipsos MORI, Health Research Authority and Human Tissue Authority	'Dialogue with public and scientist stakeholders about consent to use human tissue and linked health data in health research' (2017)	Consent; Public attitudes
12	Involve, The Carnegie UK Trust, and Understanding Patient Data	'Data for Public Benefit: Balancing the risks and benefits of data sharing' (2018)	Health data
13	Ipsos MORI and Academy of Medical Sciences	'Public attitudes to data-driven technologies in the future of healthcare' (forthcoming, 2018)	Health data; Public attitudes
14	Ipsos MORI and The Wellcome Trust	'The One-Way Mirror: Public attitudes to commercial access to health data' (2016)	Health data; Public attitudes
15	Nuffield Council on Bioethics	'Genome sequencing of babies' (2018)	Ethics; Law
16	Nuffield Council on Bioethics	'The collection, linking and use of data in biomedical research and health care: ethical issues' (2015)	Ethics; Health data
17	Nuffield Trust	'The NHS at 70: Are we expecting too much from the NHS?' (2018)	NHS capacity; Public attitudes
18	Nuffield Trust	'The NHS at 70: What will new technology mean for the NHS and its patients?' (2018)	Health data; NHS capacity

19	PHG Foundation and Genomics England	'A conversation with clinicians: shaping the implementation of genomics in mainstream medicine' (2016)	NHS capacity
20	Progress Education Trust	'What Does Consent Mean for Generation Genome?' Report for Genomics England (2017)	Consent
21	Randall Fox	'Genomics England: 100,000 Genomes Project, Potential Participant Literature: Research Report' (2014)	Public attitudes
22	Solutions Research	'Research on Genomic Data Sharing: Qualitative research report' (2014)	Ethics; Health data
23	S Skyers et al	'Count Me In! Informing the future of personalised medicine from bench to bedside' (2017)	BAME participation
24	S Skyers	'100,000 Genomes Report: Black African and Black Caribbean Communities – a qualitative exploration of views on participation' (2018)	BAME participation
25	Understanding Patient Data	'Public Attitudes to Patient Data Use: A summary of existing research' (2018)	Public attitudes
26	P Weaver and J Fitzmaurice	'Does it matter who funds science? A programme of public engagement conducted by the British Science Association for Genomics England' (2018)	Funding; Public attitudes

Methodology

The Ipsos MORI team created an Excel analysis sheet to code document content into relevant themes, such as 'Awareness and support of genomic medicine' and 'Views on the subject of a social contract'. The team then summarised these themes and added specific implications for the design of the dialogue.

The primary research papers cited in this review tend to give results which build on findings of earlier work. Therefore in this document we try to cite the most recent literature and indicate where there is consensus or difference of views across reports. Some reports are in themselves a summary of other findings; we have not referenced summaries of research which reify the contents of the original research documents they summarise, but tried to cite the original research.

2. Findings in detail

2.1 Awareness of genomics is low

Realising the potential of genomics for society will depend on the effective use of large and varied genomic databases (9). Therefore, the willingness of the public to provide data will be crucial to the success of genomics, and their attitudes are important (5). Across the studies we reviewed, many authors noted that it will be important to **monitor public awareness of, and favourability towards, genomics.**

Currently, **public awareness of the NHS genomics programme is low**: according to a 2017 public opinion poll by the Health Research Authority and National Institute for Health Research, only 25% of the population are aware of the programme (25).

We could assume that awareness is **higher among patients**: the patients interviewed by the Genetic Alliance (5,6,7) were aware of and supportive of the 100,000 Genomes project. Though the sample sizes were small and the respondents were self-selecting², their views suggest that familiarity might lead to favourability more generally. Cancer patients interviewed by the Genetic Alliance emphasised the need to explain the relationship between cancer and genomics more expressly, indicating that greater knowledge would lead to greater support (4). Indeed, more robust, representative survey data has found that those who have personal experience of genomics and consider their genomic data as ‘special’ compared to other forms of health data, are more comfortable about sharing their genomic data (Anna Middleton, personal communication 4th September 2018).

Communicating what genomics can achieve was a theme in the studies we reviewed. Some studies suggest that if there is “over-hype” about patient outcomes from genomic medicine or research, public and patients involved might become disillusioned (19), given that clinical use of genomics is in its infancy (13). The Genetic Alliance studies recommend **clarity on what genomics can do**, and how it does it (5, 6,7).

Studies also show that there are **relevant underpinning concepts which need to be explained** to the general public in order to have a fruitful discussion of the core ideas (2). Areas where awareness is limited include: understanding of the terminology and processes of data science; and the potential uses of ‘Big Data’ in healthcare overall (2, 11, 13, 14). This theme is an important one, also substantiated by public views in related dialogues for example Ipsos MORI’s work on [data science for the Cabinet Office](#) and [Machine Learning for the Royal Society](#). Understanding Patient Data has created [recommendations on vocabulary](#) after reviewing this and other work, and this dialogue should build on this knowledge.

The ‘Socialising the genome’ research and communications studies, however, have gone some way to identify what people know about genomics. The researchers point out that that people have a limited understanding of genomics and related concepts such as genes. (5)

Our qualitative impression is also that the public’s understanding of concepts such as **genes and the genome** has increased over the past 10 years. The dialogues included in this review (e.g. 11 and 13) included participants who came to the discussion with a more informed start point than we have found in dialogues of the past on similar subjects. For example, in dialogue on [Animals Containing Human Materials \(2010\)](#), or our qualitative work as part of Public Understanding of Science (2014), facilitators needed to spend a morning explaining what genes were, to everyone. In the dialogue for the [Babraham Institute](#) (2015) we needed to explain more to older participants than younger ones. In the [HRA study](#) of 2017 (11) participants confidently told us they had

² The Respondents are an unrepresentative sample. They are patients who are so motivated by their child’s condition that they have joined a support group and added to that, so motivated that they take part in a survey.

learned about this at school, or their children had learned and told them, or they were informed by the media; a contrast to the sorts of things we have heard over the last decade.

The studies and papers reviewed assume the premise that **it is desirable for the public to both understand, and contribute to, the overarching project of genomics.** The dialogue should test this assumption; what do the public think will happen if genomics is not rolled out?

There will also be a need for the dialogue to explore which approaches to communication work best; which we discuss at 2.7 below.

Implications for dialogue:

- Ensure that fundamental concepts around data, science, research, and the healthcare system today are understood. Discussions can then be firmly predicated on these concepts.
- Discuss what genomics can and cannot achieve, and what will happen if public support is absent.
- Use UPD terminology.

2.2 Different audiences have different concerns

In UPD's summary of previous research, it is noted that **younger people** are more familiar with 'Big Data' and could more easily think of benefits to using healthcare data than older people (25). We find this impressionistically true from our experience of speaking to the public during Ipsos MORI's own primary studies (e.g. 11, 13, 14, plus the Royal Society and Cabinet Office dialogues mentioned above, plus [Ipsos MORI's dialogues and Summit for DeepMind Health](#) on the principles which should inform NHS and commercial partnerships using new technology. Younger people are also disproportionately likely to be affected by genomics through their life course (15).

Awareness of and support for genomics programmes is **especially low among ethnic minorities.** In the UK, some ethnic minority groups appear unaware of genetic cancer services (1). Certain barriers to access and acceptance have been identified in US-based research, for example anticipation of negative results: African Americans at higher risk of breast cancer may refuse genetic counselling out of a fear of a cancer diagnosis. UK research concludes there may be barriers to participation for BAME groups in lack of awareness and lack of trust in the process (2). This may derive from a lack of knowledge about scientific processes for example the structure of clinical trials (23).

Analysis of data collected in a series of representative surveys across the UK, US, and Canada found that BAME groups are typically less likely to donate their DNA because they are a group who are more suspicious of official systems. In other words, they are more likely than those willing or unsure about donation to report concern about the government or police knowing something about them that they hadn't chosen to share (Anna Middleton, personal communication, 4th September 2018).

There may be **less trust** in the medical profession among BAME audiences. In UPD's summary, 26% of BAME would feel confident in a doctor's advice to participate in health research, compared with 37% white. On the other hand, the picture is more complex than an overall fear or mistrust of medical advance. Qualitative research into the literature of the 100,000 Genomes Project (21) found that BAME participants, especially those on low incomes, were *more* positive than others about the benefits of medical advances overall.

Intersectional demographic factors are present: concerns about confidentiality were raised in particular among women of African descent with lower education attainment and net income than US-born women (1). Acceptance of free breast cancer genetic risk assessment was greatest in educated women with a family history of breast cancer and relatively lower in African Americans with less educational opportunity.

We discovered very little existing research on differences around attitudes to genomics in the **devolved administrations**, so were not able to cover this theme in our review. The Genetic Alliance recommends a joined-up approach to genomics across the UK, to enable those with rare diseases to get the best access to care (6). While the current study will allow us to cover different regions in England, we recommend that the devolved administrations could be investigated in a future project.

Implications for dialogue

- Convey patient perspectives in the materials for the dialogue, rather than sampling for patients specifically.
- Use BAME and intersectional case study examples within the dialogue to ensure that we reflect a diversity of viewpoints; and ensure that our dialogue is reflective of different ethnic groups.
- We should discuss skewing sample towards younger.

2.3 Risks and benefits of genomics are identifiable; but there are also uncertainties which the public will need to weigh up

The review identifies the social and individual **goods**, and the **risks**, which are perceived to come from an individual's participation in genomics (either as a research participant or as a patient receiving treatment).

A report from Involve, The Carnegie UK Trust and Understanding Patient Data (12) provides a framework for evaluating the public benefit of any data sharing initiatives (12) (*framework summarised right*)

The factors in this framework could be applied to evaluating the success of genomics, and used in the dialogue to help participants evaluate how the data sharing aspects of a genomics rollout could be achieved.

The report recommends deeper discussion with the public of all the different kinds of benefits and risks around data sharing, in the light of this structured framework.

5 key features that a data sharing initiative designed to deliver public benefits should be able to demonstrate:

1. Enables high quality service delivery which produces better outcomes for people, enhancing their wellbeing.
2. Delivers positive outcomes for the wider public, not just individuals.
3. Uses data in ways that respect the individual, not just in the method of sharing but also in principle.
4. Represents, and supports, the effective use of public resources (money, time, staff) to enable the delivery of what people need/want from public services.
5. Benefits that are tangible, recognised and valued by service providers and the wider public.

Benefits

The studies with patients in this review identify **direct individual benefits**:

- An impressive and positive new medicine; the public recognise that genomic testing for early diagnosis and pre-emptive treatment can lead to **more effective treatment** and care, potentially improving chances of survival (20). The Wellcome Trust study on attitudes to health data (14) identifies that projects aiming for more effective treatment ("cures") is the most highly motivating reason that the public see for accepting new science. This allows them to accept potentially troubling trade-offs (such as sharing data with commercial organisations). Therefore we might expect that perceiving the translational benefits of genomics will be key to public acceptance. We should also be mindful of the fact that the public can have a simplistic view of how scientific research and translation work in practice. This was revealed in a dialogue for the Academy of Medical Sciences on [medical evidence](#), and in Ipsos MORI's work exploring how public views can influence strategy in basic research for [BBSRC](#). We should take this into account when developing materials.

- **Personalised medicine:** public dialogue participants expressed particular favourability towards the idea of tailored prescriptions, even if they lacked understanding of the medical science behind this (11).
- Patients with rare conditions express the main potential benefit as being more **accurate diagnosis** as well as effective treatment. They underlined the importance of diagnosis for conditions with which they may have lived with for years, without any concrete diagnosis or prognosis. This experience is particularly challenging for parents of children with rare conditions (7). Clinicians also believe genetics could help to reduce the length of this “diagnostic odyssey” (19). Clinicians noted that concrete benefits would be most significant for patients with rare diseases, a significant family history of particular disorders, and patients with multiple symptoms which spread across several clinical specialisms with as yet no unified diagnosis (2).
- Cancer patients see the main benefit as getting **tailored treatments** (4). Minority ethnic cancer patients are also most interested in diagnosis (1).

The expert analyses, research with clinicians, and reports of public dialogue, also establish **broader societal benefits:**

- Clinicians noted that the greater **efficiency** in treatment resulting may also lead to **reduced costs** – although the economic benefits are difficult to quantify without knowing the number of patients who will be affected (2).
- The same study of clinicians states that genomic rollout will require better data integration and data protection regulation, which could **improve overall data security** and increase the potential in other areas of medical and scientific research which make use of big data (2).
- The Nuffield Council on Bioethics’ discussion of the potential genomic sequencing of babies (15) points out that mainstreaming genomics might usher in more **awareness of, and more positive societal views of, genetic variation, disability, and poor health**. (Though the report also notes that the opposite could be true; there is debate over whether a rollout of sequencing would in fact reduce or increase genomic discrimination).
- **Building a body of knowledge in health research as a long term social good.** Creating social goods seems to be a motivating factor for the public to participate in health research generally (11). Patients are also motivated by altruism to participate in research. They want to see the benefits of genomics spread across society, and learning which will assist future generations (8).

Risks

Public attitudes to **risks around data sharing** have been covered in much of the research cited in our review, and elsewhere. The current dialogue should look at these in detail, but retain a **narrow focus on how these apply to genomics**, to avoid replicating other studies.

Risks include:-

- **Risk of unwanted re-identification of individuals:** Sharing genomic data is generally perceived by the public as more risky than the sharing of health records as it is highly personal and individual. Further concerns arise from the potential for future identification based on subsequent information later derived from genomic data (11). If a patient has a particularly rare diagnosis, there is a risk that they could be identified from this alone. The risk increases if other personal information, such as an unusual clinical characteristic, is connected to the genomic data (9). Personal repercussions could involve distress or harms due to discrimination, while research could be inhibited if the public lose trust in the process (16; and supported by all the primary dialogue research in this review).
- **Data misuse** such as cyber-security threats, state surveillance, theft or misuse of data leading to harm of individuals and institutions (16). Particular repercussions from this being genomic data could be explored in this dialogue.
- **Equity:** Public and patients in many of the studies we reviewed were concerned about a possible **postcode lottery** with respect to patient care when new technologies are rolled out. Experts suggest that equitable implementation of physician expertise and genomic technology across the more and less wealthy NHS trusts may be difficult to realise (3). Patients in particular have raised concerns about unequal access to treatment based on location or wealth (4, 6). Participants in **social groups C2DE** tend to feel more powerless to deal with consequences of personal data harms than those from other socioeconomic groups, which underlines the way that harms could bear more greatly on these social groups (25).

The Involve, Carnegie Trust and UPD report (12) also looks in detail at how to frame and weigh up risks, conceptualising three groups of risks; to **individuals, communities and public service providers**. This framing could also be useful for facilitators interrogating materials about risk.

Uncertainties

The role of **uncertainty and open-endedness** in genomic medicine is acknowledged as important in any discussion of genomics and ethics.³ This review reveals various ways in which encountering uncertainty might affect the perceptions of the public. For example, in deliberations on consent around participation in genomic research, and on access to health data, participants expressed concern about the lack of certainty about the full extent of what might be possible with shared or linked health data in general (11, 14); and said that this uncertainty might prevent them sharing their own data.

³ As set out in Montgomery et al, Ethics and the Social Contract for Genomics, ch. 16, Generation Genome

The paper by Dheensa, Lucassen et al on the challenges facing hybrid clinical and research genomics practice (3) asserted that sometimes, members of the public agree to participate in research projects with broad research purposes. Then, if they are given *too many* specifics of what the research involves, they then tend to withdraw support; perhaps this is because they become overwhelmed with detail and more aware of uncertainty.

A seminar of specialists also asked whether valid public consent could actually be possible currently, given that some risks and benefits of genomics are simply not known (20).

Key uncertainties raised in this review:

- **Scope of impact of genomics:** Genomic research is in its **early stages**, and has so far not offered significant contributions towards the understanding of common forms of many diseases (18). There are currently very few treatments which aim to change genes to prevent rare diseases, and the ones that do exist are only relevant to specific diseases affecting a very small number of people (18).
- **Diagnosis without certainty of treatment:** Genomic diagnoses may not offer sufficient evidence to classify findings as benign or disease-causing: detecting a '**variant of uncertain significance**' (20). In public dialogues, concerns have been raised by participants that this could lead to more, not less, uncertainty for patients about future cancer risks, causing additional stress (11). It could lead to patients undergoing unnecessary surgery (20). If genome sequencing of babies becomes the norm, there is a risk of over diagnosis, and preventative treatment which may carry its own risk (15).
- **Speed of uptake:** Clinicians acknowledge the **complexity** involved in mainstreaming genomic medicine. There are significant challenges around understanding the clinical applicability of genomic medicine. These include: cost-effectiveness; commissioning budgets; the challenge of physician education in a new branch of medicine; regulatory and ethical issues (19). Research with clinicians therefore recommends that public expectations about the speed of uptake and an extended lead time must be managed.
- **Longer-term changes to the fabric of society:** Social stratification, as a result of genomic knowledge being used in administrative or commercial processes could be harmful both to the individual and to society (13), but could also yield administrative and other benefits. Some potential uses of genomic data could include **crime detection, border control, uses in insurance, and employment screening** (15); all of which could create both risks and benefits, but at present are seen as uncertainties. The issue of uncertainty has been explored to some extent in dialogue (11, 13, 14). Participants tend to need prompting to think through the implications of these ideas, but when they do, are very keen not to disadvantage either individuals or whole classes of people (13).

Overall, this review suggests that research done up until now has not systematically explored these facets of uncertainty with the public.

Implications for dialogue

- Explore and compare **benefits and risks**. We suggest doing this via case studies or future scenarios which illustrate the outcomes of different benefits and risks playing out – the public can then react to these. We would also suggest ranking exercises for benefits and risks, to try and find a settlement between long and short term, public and individual risks and benefits.
- Systematically explore attitudes to the **impact of uncertainty**, to see how far this influences acceptability of genomics; establish how this affects judgments of risks and benefits.
- Draw out differences between different **socioeconomic** and other groups.

2.4 Specific issues

Incidental findings

The literature in this review highlights an important area of debate in genomics, around the role of **opportunistic screening** and the way **incidental findings** should be handled (19). While some work with the public looks at incidental findings (11, 14) there is scope to focus more on this in public attitudes work.

In [GenomEthics](#), a large-scale quantitative study including over 7000 participants internationally, 98 per cent say they want to be informed if researchers using their genetic data stumble upon indicators of a serious preventable or treatable disease.

In dialogue on the role of new tech in healthcare (13), participants felt that they would be happy for clinicians to use their existing ethical judgement on whether and how to communicate incidental findings. They were uncertain whether or not they would want to know about additional conditions *for which there is no effective treatment* (11). The Genetic Alliance calls this the “**received wisdom that people do not want to hear about things they can do nothing about**” (6).

Their studies with patients, though, on genomics specifically, shows that the patients they surveyed (cancer and rare diseases) **wish to be informed of anything a geneticist might accidentally discover** – which goes against this received wisdom. Over 50% of patients would be happy for opportunistic screening which might find out that they have an untreatable life-threatening condition (6).

This could relate to the motivations of patients to reduce uncertainty (with which they live as part of their conditions) – even if more information does not actually lead to improved outcomes.

Our new dialogue could explore these issues more fully with a wider public, deepening the discussion by looking at particular facets in detail:

- **Legal implications** of finding out additional information; the dialogue could explore the point that there is not a great deal of case law yet about how to assess how actionable results from genomic testing are (20).
- **Timing**: patients in particular cite the importance of timing in communications. This is to do with giving news at the right moment to inform and not overwhelm patients, but also about giving information with implications for care or future life (4). What impact does timing have on the wish to be informed?

An important theme in incidental findings is around **impacts on the family**. The following areas of debate could be usefully explored in dialogue:

- **Ancestry**: finding out unexpected things about your own or others’ family relationships; the ethics of using genomic data to find out non-genomic information (20).
- **Finding out information about babies and young people**. Reluctance among parents to inform children for fear of traumatising the child has been raised as a limiting factor to parents’ uptake of genomic medicine (21). There appears to be a general consensus within the international medical genetics community that only information about

childhood conditions should be shared with parents following whole genome sequencing of sick babies. However, views on which childhood conditions should be included in the information given to parents vary between countries. Practice in the UK and Europe tends towards a more focused approach to specifically determine the cause of a child's current illness. However, parents of children participating in the UK's 100,000 Genomes Project can opt to find out whether their child has several additional gene changes that can cause childhood conditions, as well as those relating to their existing condition. The British Society for Genetic Medicine have expressed concerns about this becoming mainstream NHS practice without full evaluation of the consequences for the children involved (15).

- **The ethics of finding out health information about other members of your family:** 70% of patients would want to undergo genetics tests and share their data if it would improve their own treatment tailoring, regardless of the implications on future health information about their families (4). One quarter of the same survey sample would undergo genetics tests explicitly *because* it might offer family members information on their own health (4). However, family members may not wish to know their genetic information, and a donor's discovery of a propensity to a particular disease could cause distress (15). Further, a distinct feature of genomic data from other health data is the information it provides on a donor's family. When asked about the ethical implications of family data, health representatives noted that comparing patients' genome sequences with those of their close relatives could improve clinical interpretation, and therefore restricting the 100,000 Genomes project to patients with relatives who were willing to participate could improve the scientific robustness of the study. However, the clinical aspect of the project made this selection unethical as it could lead to denial of care based on family structure (3).
- **Exploring underlying assumptions about kinship and family.** Discomfort with the familial implications of genetic results may reflect particular cultural preferences of different kinship systems. Familial interdependence – not considering an individual as separate from their family – has been associated with African American women having more negative attitudes towards genetic testing in the US (1). Kinship systems may affect the way people view inheritance due to concepts of family privacy (1). This aspect of genomic medicine may deter certain populations from participating and therefore may lead to unequal uptake across ethnic groups.

Implications for dialogue

- When exploring 'red lines' around incidental findings, include examples which allow us to discuss the legal context, the timing of information, as well as issues around ancestry, babies and young people, the ethics of finding out information about families for both individuals and society, and views of different ethnic groups.

Consent

Stakeholders and experts have considered issues around consent (20) and public and patient views have also been canvassed (11, 4, 6). The HRA's recent dialogue (11) covered the mechanics of consent in some detail. The Genetic Alliance's patient research (4, 6) also asked many questions on consent, ultimately recommending dynamic consent where possible for genomics.

Despite this coverage, there remain significant debates over the best way to consent participants in genomics (5). These issues could be discussed further with the public in the light of a genomics rollout, such as:

- Ethics of **automatic opt-in for all**, in terms of the social contract.
- **How equitable dynamic consent can be**, given varying levels of digital literacy.
- **The ethics of potential future uses of data**: The report from the Nuffield Council on Bioethics (15) points out that as genomic and data mining techniques improve, existing data may yield more and more insight. This could occur to the point where the individual could not have been reasonably expected to foresee how their data could be used when they originally consented (15). A [recent study by the National Data Guardian](#) tests the idea of "reasonable expectation" when weighing up consent issues. They conclude that this concept is a difficult one for the public to think about – even across a three-day event – and that participants tended to gravitate towards talking about whether they supported the purpose of the data sharing or not, rather than abstract formulations about reasonableness. The current dialogue will need to find new ways to explore this.
- **The role of counselling** and trained advisors (11, 5)

Implications for dialogue

- Explore public attitudes to consent in the light of the uncertainties of future uses of data, and find new ways to bring to life "reasonable consent". Explore dynamic consent in the light of this.
- Explore the appeal of automatic opt-in and its benefits and drawbacks in the light of the social contract, and the role of counselling.
- Do not cover consent processes in detail, this has been covered in previous studies.

2.5 Views on access to, and use of, genomic data

Who should have access?

Access to healthcare data has been a hot topic in recent years. Research and dialogue with the public has explored this in detail (12, 14, 3). Bodies in the public sector have wanted to understand the extent of any public mandate for data sharing across both public and private sectors, while both the NHS and private sector are increasingly keen to avoid reputational risks around data protection caused by sharing data too recklessly⁴. This has informed various public dialogues and a range of other studies.

The Wellcome Trust study (14) discovers that the public tacitly apply four tests when the question of access to healthcare data by commercial organisations is considered. The degree to which the public think that commercial access to their health data is acceptable is subject to **conditions about how the data will be used**. The 'Why' being the most important question; public benefit must be a core aim of the data sharing. This also fits with the Involve/Carnegie Trust/UPD framework (12) where 'purpose' is a key factor in evaluating the success of data sharing.

It seems this process of evaluation applies to genomics. Reports show that people are willing to accept that the private sector can have a significant role in developing treatments for rare disease – even surpassing the role of the public sector, and even if they may have been initially doubtful about the positive role of commercial companies (21). If participants have information about how the data will be used and who will have access to it, they will be more willing to accept the use of their data (21).

The public tend to trust the motivations of clinicians – who are perceived to prioritise immediate care of their patients – more than researchers, whose motivations they see as varied (11, 14). Patients and public are also more suspicious of industry-funded scientists than publicly-funded scientists, as they fear the former may favour financial gain over public welfare (20).

The Genomics Conversation report notes that many people are eager to share their data if there is a benefit to society, but are less eager when profit-making companies are involved. Research by companies who profit from the sharing of genomic data is only considered acceptable if it brings about a wider social benefit (5).

The current dialogue will not need to explore the different circumstances in which commercial access is acceptable in general, as this has been covered so thoroughly.

Instead we should focus on the **detail of the 'red lines'** held by the public about genomics specifically. To avoid replicating what has been done already, the dialogue could cover:

- **Specific risks to individuals and society around insurance uses:** In the BSA Futures Debates, (5) over 95% of respondents reported they would be unhappy to share genomic data with insurance companies. The most common fear expressed by public and patients is that health insurers will use genetic information to create stratified premiums or limit access to

⁴ Particularly in the wake of well-known data privacy examples such as the Information Commissioner's ruling that DeepMind should not have had access to patient data given by the Royal Free Hospital. <https://www.theguardian.com/technology/2017/jul/03/google-deepmind-16m-patient-royal-free-deal-data-protection-act>

insurance entirely (13, 4, 6, 11, 13, 14). This could create harms for individuals, plus there are concerns about the implications of a national genomic database on society generally (9). In the HRA's recent dialogue (11) participants felt that the current moratorium on data sharing with insurance was not a reassurance, as this could be revoked in future; and they felt that genomic information once shared cannot be retrieved. This is part of broader concerns about sharing personal data with the private sector (12, 14), but goes beyond this to being a specific concern. There are particular concerns from BAME communities around insurance uses (22,24) based on historical experiences and narratives around ill-treatment and discrimination towards BAME groups in medical research. There are concerns to the point that financial discrimination could lead to a form of social eugenics.

- **The nature of public and private partnerships** which may underlie the realisation of genomics. The dialogue should include discussion of what the public want the role of private genomic services to be in the healthcare system, and how it relates to the work of the NHS. This could include more explicit discussion of funding and how to optimise the value of health data; including some of the options for different data ownership models which have been proposed⁵, but not explored with the public as yet. We should discuss the implications if the NHS cannot provide all services to all, and then private providers might deliver commercial services which are also unequally distributed, or may include ethical or technical strictures on their products and services which do not meet the high standards of the NHS. 90 per cent of people support the founding principles of the NHS, indicating that these principles are just as relevant today as when the NHS was established ([King's Fund and Ipsos MORI](#), 2018). Genomics England is also trusted to have participants' interests at heart (8). We will need to cover public expectations of the bodies involved and the way the social contract should cover new relationships. Overall, we could ask; who should a new social contract include? The NHS and the public, but what might be the role of other bodies?
- Discussion of **international data sharing of genomic data in particular**. While health data has been shared across many boundaries for healthcare evaluation, and is broadly acceptable, "we are now entering a new era of connectivity, with plans to link entire health systems, across countries, to each other." (9)
- Anna Middleton (9) also notes the principles enshrined in the International Declaration on Human Genetic Data, among which is the need for adequate privacy protection. The nature of that **privacy protection** and the safeguards which can be expected should be discussed. The question of who should have the 'decision power' to make judgements about data access is also a live one for the public, discussed in the BSA deliberative dialogue on genomics (5). This could be further explored in the current dialogue.

⁵ For example a biotrust model, in this paper <http://www.lancaster.ac.uk/fss/journals/gsp/docs/volume1number3/dwgspvol1no32005.pdf>

Differences between different groups

Young people are more aware of data security issues than older, and 55% (18-34 yo) compared to 62% (55+ yo) are concerned about data security (25,5). Public acceptability of commercial access to health data also appears to be linked to demographic factors: educational attainment, social grade, and broader understanding of how data is used affected correlate with greater acceptability (14).

Public and patients see their health data as having **value to society** (4, 6, 11, 14) – though people in groups ABC1 are more likely than those in C2DE groups to view health data as having potential benefit to society (25); hence there may be socioeconomic differences around perceptions of value.

Implications for dialogue

- Dialogue should include discussion of insurance, private and public partnerships, international sharing, and explore hypotheses around different data relationships and roles of different providers.
- Specifically on insurance, while we already know that the public are concerned about insurance uses of genomic data, there is scope for more granularity on what they are most concerned about.
- We should keep a tight focus on genomics, as these issues have been covered already around health data generally.
- We should split out findings by different groups.

2.6 Current and future capabilities of the NHS

Views on the health of the NHS and its readiness for genomics will be relevant background for this project.

In the Genetic Alliance studies, risks and benefits are assessed by patients based on how well learning from genomic data is **translated into care**, and how effectively research can be passed through to clinical practice (6). Patients, public and clinicians have also noted that the benefits of genomic medicine are dependent on the **turnaround time** of results and how they are used clinically (2, 4).

However, research with clinicians reveals that they feel the health service is **'a million miles' away from being able to deliver genomic medicine** (19). A survey in Canada with medical oncologists involved in genomics trials identifies that most do not see themselves as experts in genomics, especially when it comes to newer techniques (2). The PHG and Genomics England held an event with the All Party Parliamentary Group on Personalised medicine, at which parliamentarian attendees wanted to focus on NHS capacity, including the number of pathologists needed (5).

There is, however considerable **public trust in** the NHS. The public tend to assume that health research is conducted under the auspices of the NHS, and have limited knowledge of other actors

involved. This leads to assumptions that all health data is handled under with clear and consistent safeguarding protocols, is never used with a profit motive in mind, and that accountability is straightforward: leading to implicit trust in the system as it is (11), and potential risks for a future genomics rollout which might include more complex processes.

This dialogue could cover public aspirations and concerns about the following areas, which have not been covered in depth:

- **How a hybrid approach to research and clinical care should be deployed in genomics:** Research and clinical care include different models of participation and feedback. Thinking about the hybrid nature of genomics raises questions about different thresholds for understanding participation, and discussion of practice from clinical and research (3). Public and patients know that findings from research may not yield them personal information or benefits (8, 11); but at the same time the Genetic Alliance recommends that the pathway from research to care should be as streamlined as possible (6). If the direction of travel is towards personalised medicine, equity could be hard to maintain as treatments appropriate for a diversity of patients with different genetic mutations may not exist. The NHS must develop systems for how to manage this, particularly with respect to diseases with a higher association among different ethnic groups (10). There are other issues, such as how the NHS might benefit from population-wide screening, and what the approach should be to genetic counselling, which could be discussed here. (6).
- **How should we fund a rollout?** The Genetic Alliance recommend in their studies (4, 6) that for benefits to be realised, the whole NHS workforce will need to be upskilled to manage genomic data and its implications. The public currently are very concerned about preserving the NHS (healthcare and the NHS are seen as the most important issues facing the country, second only currently to Brexit in the [Ipsos MORI Issues Index](#)).
- **How can the NHS demonstrate its data protection capabilities?** In the AMS study on future technologies in health participants questioned whether the NHS has the capacity to work with future tech providers, negotiating partnership and explaining relationships to the public. The NHS bears the weight of responsibility for safeguarding data and for doing this well (13). This could be discussed in more detail.

Implications for dialogue

- Explore; how can genomics be rolled out given the current strengths and weaknesses of the NHS?
- Explore what the public would like to see in terms of readiness for genomic rollout on the clinical practice side.
- Explore the different relationships around data which the NHS might have to oversee, and what the public expects the NHS to demonstrate in terms of its capacity to protect data.

2.7 Communications around a wider genomic rollout

Genomics England is required to bridge the divide between the way the public talk about genomics, and the way academics and clinicians might discuss the issues. (5) The 'Socialising the genome' engagement project funded by Genomics England, the Wellcome Trust and the Wellcome Sanger Institute and the work of Understanding Patient Data generally, have explored the language and concepts which would best cut through with the public on genomics, and this dialogue will need to build on the good principles already developed.

The patients researched by the Genetic Alliance considered it quite easy to explain the concept of genomic sequencing (4). This was a self-selecting group of people who already knew about the process in detail, but it implies that once involved in the project, the concepts are not too difficult for people to grasp.

We should report on how best to communicate with the public **around a wider rollout**. From the review of literature, the following areas are likely to be important:

- **How to increase awareness of genome sequencing and the 100,000 Genomes project.** Participants in public dialogue workshops have expressed genuine surprise that this technology existed, and that it could benefit patients with a range of health conditions, including infectious diseases, and not just those who have a rare disease (11). How should the capabilities and limitations of genomics be conveyed?
- **How and why genomic information might be shared,** and the potential complexity of the relationships between the NHS and other bodies.
- **How do the public respond to the communication of uncertainty?** Once aware of the project, patient expectations must be carefully managed – especially in circumstances where tests are unavailable or ambiguous (10). Diagnostics must not be overpromised: participants must understand that identification of disease susceptibility will not always remove the risk of that disease (10). Even where drugs are developed based on genomic findings, they may be very expensive and may not be approved by NICE (10). Public expectations may be more realistic if there is a media effort to convey the uncertainties in this field and deliver a less deterministic message on the potential for diagnoses for research participants (20).
- **How to balance benefit and risk:** Participants in dialogue on medical innovation emphasised that although a good understanding of risk was required to ensure informed consent, an over-emphasis on risks can lead to the benefits being forgotten. It was suggested that communications should focus on the risk–benefit balance (10).
- **Too much information or too little?** Information should be sufficiently concise that participants find it easy to digest. Research shows that participants of the 100,000 Genomes Project do not always read the information provided (3). Providing further information may cause confusion or a feeling of being overwhelmed. This needs to be balanced against the need for informed consent.

The needs of particular groups. Qualitative research on BAME patients suggests that there is some feeling that medical consultations focus too heavily on the collection of information and do not take sufficient time to explain why this is useful or necessary (1).

However, pilot community-based cancer genetics services, held at venues including faith centres and cultural events, and distribution of translated leaflets to raise awareness of familial cancer may improve satisfaction with genetic services and reduce religious sensitivities when conveying bad news (1) – although in isolation these initiatives have not yet been associated with significant uptake by ethnic minorities.

Implications for dialogue

- Identify the most motivating ways to communicate around the wider rollout, covering increasing awareness of the potential of genomics, its uncertainties and risks; the level and detail of information required, and the different needs of particular groups.

For more information

Ipsos MORI 3 Thomas More Square
London
E1W 1YW

t: +44 (0)20 3059 5000

www.ipsos-mori.com

<http://twitter.com/IpsosMORI>

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