VIEWPOINT

Research participants: critical friends, agents for change

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INTRODUCTION

The Participant Panel at Genomics England

This article describes how participants in large scale genomics projects can help to shape and steer them, on behalf of wider participant communities, based on the experience of the Participant Panel at Genomics England [1]. We can be 'invited guests' and we can be 'gate-crashers'. The article identifies some transferable lessons that could be applied by those who seek to establish or develop large scale health research programmes in the future. It has been written entirely by Panel members.

Genomics England was established in 2013, with a mission to deliver the 100,000 Genomes Project in partnership with NHS England. This was to assess the value of whole genome sequencing as a mainstream health tool, for implementation in the wider NHS [2]. Initial findings have been encouraging: new diagnoses for 25% of participants with rare conditions, and 'actionable results' for around 50% of participants with cancer (adjustments to their treatment, and/or opportunities to join new trials) [3].

From the outset, Prof Dame Sally Davies (then Chief Medical Officer for England) was clear that participants must be involved in overseeing how their data was used [4].

The Participant Panel was established by Genomics England in spring 2016, with the full support of Prof Mark Caufield (then Chief Scientific Officer), Vivienne Parry (Head of Engagement) and Prof Mike Parker (founding Non-Executive Director and Chair of the Ethics Advisory Committee). People who were signing up for the 100,000 Genomes Project, and their primary carers, were invited to apply. This meant that everyone around the table had a range of experiences of NHS services, and shared the journey of consenting to have their whole genome sequenced for research purposes.

Panel members come from all over England, with a wide range of health conditions and patient journeys. About two-thirds of the Panel have experience of rare health conditions as a patient or carer. Around one-third are or have been cancer patients. We are all driven by a desire to transform the difficult situations that we have faced into a positive force for good. We want genomics to deliver answers to as many undiagnosed patients as possible, to help them access the support they need, and to drive progress against cancer. We want health data custodians to facilitate as much research as possible, while adhering to agreed safeguards.

The Panel oversees what Genomics England and its partners do with our data. We advise Genomics England about what matters to participants in genomics research and how it should shape its services to ensure that patients benefit. The Panel can invite anyone involved to discuss their work at a Panel meeting, and the Chair reports directly to the Genomics England Board.

The Panel holds quarterly meetings where Genomics England's senior leadership team are invited to share their work and engage us in discussions about strategy, participant engagement, project outcomes and next steps. With our varied life and patient experience, we bring robust viewpoints to the table, acting as a critical friend.

Panel members may claim an activity payment (in line with NIHR recommendations [5]). This is an essential element in making us feel valuable, and enabling us to step away from our regular jobs and caring responsibilities to commit to this important role. However, this payment is discretionary because of the impact that extra earnings can have on an individual's eligibility for existing state benefits [6].

Genomics England provides a secretariat to the Panel which enables us to engage in diverse initiatives, public dialogues and contribute to its core committees (described below). Over the years our workload has grown in parallel with Genomics England's remit, which brings both challenges and opportunities. However, we exist as an advisory team, not a delivery team. This is a critical distinction that allows us to retain our independence.

The Participant Panel is acknowledged as a key contributor in delivering the UK Government's genomics implementation plan [7].

PARTICIPANTS AND THEIR FAMILIES CAN CONTRIBUTE TO GENOMICS RESEARCH

Note that many other organisations have already been writing about this subject. For example, the Global Alliance for Genomics and Health (GA4GH) have produced a Regulatory and Ethics Toolkit which includes a helpful framework [8].

In our experience, participants and their representatives can get involved in many different research activities. We can see two main routes:

Invited to the party – contributing to initiatives that are led by data custodians or researchers; and
Gate-crashers – participants as agents for change: identifying and catalysing initiatives that are important to us and the communities we represent.

Invited to the party

Decision-making. At the most fundamental level, research participants should have a place at the table when decisions are

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being made about how their data will be used. At Genomics England, Participant Panel members sit on the Ethics Advisory Committee [9], the Access Review Committee [10] and the GECIP Board [11], actively deciding or advising how our data should be used. We see this as an essential role. It ensures that decisions about health data remain close to the values of the people whose data is involved. It helps the wider participant community to have confidence that Genomics England has their interests at heart. Panel members learn a lot through their interactions with scientific and clinical leads who they would otherwise never meet. It takes a certain degree of assertiveness and self-belief to make contributions in such company, and it has not always felt as though the participant perspective was unreservedly welcome. However, this is the point of having diverse viewpoints around the table: it makes the overall deliberation much more robust and delivers final decisions that are better-informed.

The Panel has also contributed to several deliberative discussions about complex genomic questions, that in turn helped to shape public policy. These include a study in 2018–19 exploring public aspirations, concerns, and expectations about the development of genomic medicine in the UK [12] and more recently we have been helping to shape the dialogue around the Newborn Genomes Programme [13].

Communications. Participants and their families can advise researchers on the best ways to communicate with the wider participant population. In genomics, these messages can be life-changing, and we know from experience that getting the communication wrong can have devastating long-term impacts.

The Panel are often asked to help prepare and disseminate material about Genomics England's work. We review and contribute to website content and advise on accessibility. We appear in the media to tell our stories and share Genomics England's work and patient benefit. Several Panel members have also shared their perspectives on the *G Word* podcast series [14].

This is a very traditional role for participant representatives, but it must not be the end of their involvement. We move beyond simply acting as the 'shop window' (advertising the scheme to others) and are committed to 'getting behind the counter', to have a direct impact on the direction of genomics research itself.

Research implementation. Genomics researchers who intend to deliver results to individual participants need to test that the participant recruitment and results delivery pathways are functioning as expected. Participant representatives can add a lot of value by reporting their experiences back to the people running the scheme, in real time, and directly show them how their intentions can differ from reality.

The Panel helped to identify such issues in the 100,000 Genomes Project, and pushed for the development of a 'track my sample' service so that participants awaiting their initial results could find out where they were in the queue. Around 10,750 participants have used this service to date. We believe this has been reassuring for participants, many of whom were hoping for results that could change their life, that they had not been forgotten and were still due to receive a result in due course. This also had an NHS benefit as it reduced the amount of clinician time spent trying to find out information for their patients.

Sometimes research participants' voices can help lend weight to the research endeavours themselves. Despite prior ethical approval, researchers sometimes struggle to access the data they require for analysis. Representation to data custodians by participants, reminding them that consent for use of their data and samples has already been given, can unlock these research log-jams.

New ethical issues. Participants can also advise on ethical matters which could not have been foreseen at the outset. Involving participants in deciding how to proceed helps to ensure that the research continues to meet their expectations, or will adjust in ways that are most acceptable to the community.

For example, the success of the 100,000 Genomes Project means that it has to tackle the challenge of re-consenting young participants when they reach adulthood. These young people need to decide whether or not they wish to continue to participate as an adult (if they have the capacity to do so). Working out how to approach these families and young people was not straightforward: everyone's circumstances differ [15]. Our perspectives as parents of children with widely varying levels of cognitive ability helped to shape Genomics England's approach.

Observations. All these examples rely on the lead researchers to take the initiative, to reach out to participant and patient communities, and to be open to discussions about what matters to them. We would always encourage this, with caution; some of these discussions can bring up traumatic experiences for patients or their families.

Many genomics research participants live with rare or chronic health conditions; their time and energy are especially precious. Being clear about what scope they have to shape the research – and where the limits are – will help to set realistic expectations for all involved [16].

Project / organisational leaders should consider carefully from the outset who needs to be involved, and how relationships need to be structured. Where does the real power lie? Who gets to choose what the participant representatives do? How will their time be funded? What support do they need from the professionals to 'get up to speed' and understand the context for the project/organisation?

Success relies on developing a strong and sustained relationship between the participants and the project leadership, and a culture of partnership, openness and reciprocity. Once there is a strong foundation, and an ambitious group of research participants, things can get really interesting – as we will now explain.

Gate-crashers

Genuinely empowering research participants can sometimes lead in unexpected directions, but these can add significant value to what was originally proposed – creating benefit for the participant community as well as the project leaders.

Additional findings. Everyone who signed up for the 100,000 Genomes Project was offered the opportunity to hear about 'Additional Findings'. These are genetic markers indicating a likely predisposition to certain forms of cancer or other chronic conditions [17], for which risk-reducing measures and/or treatment are available in routine NHS care. Around 82% of the participants opted to find out their Additional Findings. The initial plan was to only contact the participants who had positive findings (i.e. an 'actionable' result), but the Panel pushed hard to make sure that everybody who requested Additional Findings would receive a result, no matter whether these were positive or negative.

For many reasons, the gap between signing up and receiving answers from the 100,000 Genomes Project grew to span several years. By the time Genomics England was ready to begin looking for Additional Findings, we were concerned that many participants may not have remembered whether they asked for these or not, or their circumstances may have changed so they might not receive their result. Post Covid, we were also aware that people's appetite for further health knowledge may have changed, and many participants with rare conditions and cancer were already grappling with significant and changing health issues. We secured the agreement of Genomics England to work with its NHS partners to write to every participant in the 100,000 Genomes Project to remind them about the initiative, and to offer them a chance to 'check their choice' before the analysis was undertaken. The Panel advised on the

Panel member, Dave McCormick has recently worked with Genomics England and three national sight loss charities to produce a webinar series, 'Sight Loss and Your Genome'. These have enabled over 100 people affected by visual impairment to hear from several of the leading researchers in the field and ask questions in an interactive Q&A session. Feedback from the second event showed evidence of participants wanting to start to influence the direction of research.

Key lessons from this case study:

• Choose an accessible technical platform in collaboration with the community you are aiming to reach – in this case, a platform that can work effectively alongside assistance technology applications, such as SupaNova or ZoomText, which support patient participants with sight loss

• Advertise the event through community support groups and social networks, starting around 1 month in advance, for maximum uptake

• Make the event engaging - for example, use a 'radio show' format instead of formal presentations and facilitate conversation between the researchers and the audience

Fig. 1 Case study: improving links between genomics researchers and patients.

wording of these letters and the overall timing of the approach. Over 76,300 letters were sent out, and the Genomics England service desk received over 12,000 responses. A large majority of the participants were reassured by the opportunity to check; just 327 people subsequently changed their choice, although a few were rectifying mistakes that had been made when their original choice was recorded.

Language and terminology guide. In early 2022, the Panel recognised that there was a gap in the information given to the many new starters arriving at Genomics England as the company pivots from a project into a service delivery company [18]. Many of these colleagues may never have worked in healthcare before, and very few have had any contact with the people whose data they are responsible for curating. Consequently, they were unsure how to talk with us or about us.

The Panel had a round table discussion about the importance of getting the language right when engaging with patients, research participants and their families. This was distilled into a Language and Terminology Guide for anyone working in genomics and healthcare. The guide was launched at the Genomics England Research Summit in London on 4 May 2022 and simultaneously shared online [19]. It has been warmly welcomed by many different actors in the genomics landscape – and beyond. Almost 1,000 hard copies have been given away so far, while the online version has been accessed over 500 times.

Position statements. Less high profile, but equally important, have been the position papers that the Panel have produced for internal circulation among Genomics England colleagues over the years. These have set out the Panel's views on matters such as the involvement of industry in genomics research, and the direction of genomics research within the organisation. In a 'many to many' relationship, where many different staff would like to reach out to many different Panel members in their various roles across the organisation, it can be an efficient way of making sure that everybody is getting a unified message.

Making links between researchers and patient communities. There are few opportunities for patients and research participants to meet the genomics researchers who use their data to make discoveries. We want to bridge that gap, to allow researchers to present their work to the patient community and enable patients and families to ask questions. See Fig. 1 for a recent example of how this can be achieved.

Embedding participant voices into the wider genomics landscape. Panel members also use their lived experiences to inform other initiatives that they hope will benefit the wider patient/participant community. The Chair of the Panel has been advising NHS England's Genomic Medicine Service (GMS) about setting up an equivalent structure to ensure that patients and their families are heard in decisions about how the GMS is run [20]. As part of this, she wrote the draft Terms of Reference for the group that has become the GMS People and Communities Forum, which brings together the leading regional and national 'Public and Patient Voices' on a quarterly basis. The Vice Chair and several other Panel members are also advising various regional GMS Alliances about how they can best embed their participant representatives in their own leadership structures.

Observations. Panel members have felt empowered to take these 'gate-crasher' actions because of the support they've received from Genomics England. But they have also moved beyond where they were invited to be, driven by their ambition to share what matters to patients and their families more widely. These initiatives have not only made Genomics England more responsive to the needs of its participants, they have helped to raise the profile of genomics among the communities who stand to benefit, including the wider public.

REFLECTIONS AND RECOMMENDATIONS

As a relatively new discipline, genomics is not fully understood by the wider public and sometimes viewed with suspicion [21]. It requires multidisciplinary working and collaboration [22], helping to create an atmosphere of discovery in which every patient voice has perhaps got more opportunity to be heard than in traditional medicine. The wider family implications of genomic discoveries mean that traditional patient-clinician relationships need to evolve [23]. The growing recognition that genomics is a team sport lends weight to the argument that more patients and their families should be involved in deciding how genomics is delivered. We are all on this team. Our DNA is literally powering the discoveries in this field and there are many ethical and practical reasons why we should speak into decisions about how scientists are using it.

Since the day it was founded, the Participant Panel has been invited to scrutinise and comment on what Genomics England has been doing with our data. The initial focus on delivering the 100,000 Genomes Project has been relatively straightforward – although we all bring different perspectives, there is one broad target to aim for. However, as the organisation has grown and diversified, the Panel has sometimes struggled to keep track of the strategy driving the new initiatives. It is now impossible for our group of volunteers to have oversight of everything.

Here are some lessons from our experience, for any large genomics research organisation:

First, that in order for participant voices to remain at the heart of an organisation, they need to echo around it. For example,

participants are an integral part of the 'Welcome Week' onboarding of new staff at Genomics England; everyone gets to hear that 'every data point has a face' [24] and the rhetoric around 'participants as the North Star' infuses corporate publications such as the Annual Report [25]. Our Language and Terminology Guide is widely shared.

Second, that specified senior members of the leadership team need to routinely seek participants' input to the organisation's activities. Few patient/participant representatives arrive with the confidence and experience to be a 'gate-crasher' from the outset; most will need time to develop relationships with each other as well as with the staff they are seeking to influence. Knowing 'whose door to knock on' is really key in becoming an effective advocate but this job is much easier where some doors are well signposted and always open.

Third, that there must be clear Terms of Reference, and realistic expectations about how much time the volunteers can give to their roles. It also helps to have a 'gate-keeper' on the staff of the organisation, working closely with the participant representatives to identify appropriate opportunities for engagement. Strategic and fundamental participant issues should always take precedence. If there are too many invitations for participant input in too wide a range of things, some individuals can become overburdened, with a resulting loss of 'job satisfaction', reduction in the quality of their output, and/or an over-emphasis on one point of view. The 'gate-keeper' can also help new arrivals to understand the technical terminology and the organisational landscape in which the research sits, both of which are essential for meaningful contributions to group discussions.

Fourth, that over time, keen 'lay' people can develop a highly specialist knowledge of the field. We are no longer as representative of 'the average' patient or participant, so the advice we offer begins to be tailored to the language and processes used by the organisation already. As one of our Panel noted, *'you can't get inside unless you absorb the culture and become a critical friend'*. Using that insider knowledge to apply pressure on the parts of the system where we see things that need improvement is good. But as time goes by, we lose our legitimacy as 'lay' people so need to be mindful of the gap. Keeping in touch with wider participant communities via our other networks is an important way to address this. We recognise the need to expand the diversity of viewpoints we represent, and support new members to learn about relevant background issues. We recently recruited our first participant from the GenOMICC study [26] and are also inviting expressions of interest from anyone who has consented to research through the GMS. If you are interested, please write to ParticipantPanel@genomicsengland.co.uk.

Fifth, that some initiatives will require additional work to bring together representatives of the communities they seek to help. Our legitimacy as representatives of the 100,000 Genomes Project participant community derives from the fact that we are all rooted in that experience ourselves. We know about cancer and rare conditions but we cannot and should not be expected to speak for others. Consequently, it is important to ensure that new voices are invited into the room when discussions about them are getting underway.

In conclusion, the Participant Panel at Genomics England is confident that it has made a significant impact on the design and structures of the organisation, to better serve the wider participant communities. As the scope of Genomics England has changed, we have too, with a flexible and pragmatic approach to the challenges and opportunities that come with further funding and additional projects. Genomics England often calls participants its 'North Star' reflecting the principle that patient-led design will result in better research and clinical outcomes. We see future opportunities for further collaboration with academic and commercial researchers using our data to progress healthcare in the UK and globally. Such initiatives must be centred around the need to build trust with patient communities, and to listen to and empower those whose data is being analysed. We are proud of our work to date and are committed to our role in building a genomics healthcare ecosystem based on trust, reciprocity and partnership with its participants.

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AUTHOR CONTRIBUTIONS

All of the authors contributed equally to the initial workshop which informed this article, reviewed it, and submitted comments. JHW convened the workshop and drafted most of the article. RM, IKG and HW made substantial contributions to the text, alongside DMc who also provided the case study (Fig. 1).

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COMPETING INTERESTS

JHW is the independent Chair of the Participant Panel at Genomics England, a member of the UK National Genomics Board, and a founding Board Member of the CureGRIN Foundation. She also sits on the NHS Digital Research Advisory Group and is a lay adviser to Health Data Research UK. RM is the independent Vice Chair of the Participant Panel at Genomics England, and Founder and CEO of Hereditary Brain Aneurysm Support (CIC). DMc is a Lay Patient Representative to the MSc in Genomic Sciences, Manchester Academy for Healthcare Scientist Education (MAHSE) Patient Forum, and Interim Chair to the Patient & Public Voice Panel, North West Genomic Medicine Service Alliance. SS is a Trustee of Disability Rights UK, a member of the PPIE Panel of the North Thames Genomic Medicine Service Alliance, a lay member of the Neuromuscular North Star Clinical Network, a campaigner for the Changing Places Consortium and the founder of Fraser & Friends (Disability Rights & Rare Disease Advocate). LC is a community paediatrician, Sirona Care & Health.

ADDITIONAL INFORMATION

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