**Fostering trust in healthcare: participants’ experiences, views, and concerns about the 100,000 genomes project**

**ABSTRACT**

In this paper, we present findings from a project involving 20 patients with rare diseases, or parents thereof, participating in the 100,000 genomes project (100kGP) about their experiences of, and views about, the project, including why they took part, and their hopes and concerns about the future of genomic medicine. Patients who attended genetic clinics for testing were offered the opportunity to undergo the more extensive whole genome sequencing (WGS) if they agreed to take part in the 100kGP. Once people had agreed, a specific additional appointment was organised for them; taking part in the project therefore involved additional travel and appointments. We found that interviewees’ decisions to participate in the 100kGP were based on interpersonal and institutional trust in the NHS, and on an investment in improving care for the future.

Interviewees relied upon receiving good ongoing NHS care for managing their own or their child’s rare disease, but they worried about what their relationships with NHS healthcare professionals would be like in future. A few participants worried about whether Genomics England’s biorepository would remain protected and an asset of the NHS. To honour and foster participants’ trust – which may easily be lost - and their ‘clinical labour’, we therefore recommend ongoing public engagement and consultation about how genomics is being integrated more widely across specialties (especially given its current funding and staffing constraints) within the newly formed NHS Genomic Medicine Service.

**INTRODUCTION**

**100,000 genomes and the genomic medicine service**

The Chief Medical Officer for England focussed her 2016 annual report on genomic medicine. Entitled ‘Generation Genome’, the report “identifies the opportunities that advances in genomic technology can deliver for clinical practice and public health”, and called for a “genomic revolution” (Davies, 2017, Ch1 p8) in the National Health Service (NHS), in which genome sequencing, and precision medicine (also referred to as stratified or personalised medicine[[1]](#footnote-1)) become a routine part of clinical care for treating rare and common conditions. The 100,000 Genomes Project (100kGP) has been laying the groundwork for an NHS genomic medicine service since it was announced in 2012. As part of the project, whole genomes from NHS patients (and family members – or tumours- thereof), with either a cancer or rare disease, are being sequenced. There are between 5000 and 8000 different types of rare diseases, including cardiological, dermatological, neurological, ophthalmological, renal, and respiratory conditions.[[2]](#footnote-2) They are individually rare, but collectively common, affecting 6-7% of the UK population. Largely, they are not diagnosable with targeted genetic tests. In the rare disease group, participants’ DNA is compared with that of family members; in the cancer group, it is compared with the DNA within their tumour. People with heritable cancers (e.g. BRCA1) were recruited within the rare disease arm.

Genomics England Limited, set up and funded by the Department of Health, has been overseeing the project with thirteen NHS Genomic Medicine Centres, comprising 70 hospital trusts across England involved in recruiting patients. From late 2018, the work of Genomics England is being subsumed within NHS England[[3]](#footnote-3) from late 2018 (Chisholm and Wordsworth, 2017) and its activity becomes part of mainstream NHS practice. The 100kGP was a deliberate hybrid of clinical practice and research: potential clinical benefit comes from the analysis of whole-genome sequence data, which might lead to, or confirm, a suspected diagnosis or prognosis of a rare disease and cancer, respectively. The sequence data linked with clinical information enter a biorepository for future research projects. Given the hybrid nature of the project the consent process is complex. Patients (or participants) do not receive detailed information about specific studies or who will do the research, which are unknown at the time the patient agrees to take part - but they will likely involve academic researchers (e.g. universities) and/or commercial researchers, who will access de-identified subsets of the biorepository data.

Figure 1 shows the process of recruiting patients, although this may have differed from one centre to another. Patients who attended genetic clinics for testing were offered the opportunity to undergo the more extensive whole genome sequencing (WGS) if they agreed to take part in the 100kGP. Once people had agreed, a specific additional appointment was organised for them; taking part in the project therefore involved additional travel and appointments. Research nurses and clinical trials assistants sought consent at the genomic medicine centre we recruited from. In other genomic medicine centres, genetic counsellors, consultants, junior doctors also sought consent.

The falling costs of sequencing made this project possible but translating the technology into useful clinical information is complex and the downstream practice implications will be expensive (Chisholm and Wordsworth 2017). At the same time, when used for the right patients (i.e. where there is evidence for potential effectiveness), genomic medicine could prevent ‘diagnostic odysseys’, improve prediction and prevention, lead to earlier diagnoses or to precision treatments that are more effective for patients and cheaper for the NHS (NHS England, 2016).

In this paper, we present findings from a project involving interviews with 100kGP participants about their experiences of, and views about, the 100kGP, including why they took part, and their hopes and concerns about the future of genomic medicine. Our findings show that interviewees decided to take part in 100kGP because their feelings towards it were bound up with their feelings towards their healthcare providers and the NHS, an institution that they trusted, relied on, cherished, and wanted to improve. At the time of writing, we are in a transition period between recruitment to the 100kGP and the new NHS Genomic Medicine Service.

**METHODS**

SD and AF, both social scientists, were not involved in delivering 100kGP. AL, a consultant in clinical genetics, referred some patients to 100kGP and sits on Genomics England’s Ethics Advisory Committee. An NHS Research Ethics Committee approved the current study.

We sent everyone who consented to WGS to investigate potential rare disease at one recruiting Genomic Medicine Centre (covering population of 3 million), an invitation to participate in an in-depth interview[[4]](#footnote-4). SD then contacted those who sent back an expression of interest to participate, or spoke to those who contacted our research office, to answer any questions, offer a telephone or face-to-face interview, and arrange a suitable time, date, and location. We initially used a convenience sampling strategy to select from willing participants, arranging interviews with those who sent back reply slips and were available for interview. Several of the first few participants happened to be aged over 65, were retired from employment and had adult children and we thereafter focused our sampling strategy on younger participants who were in work and whose children were still young, to explore the different sorts of issues raised. The interview schedule was semi-structured, with the broad aim of exploring views and experiences of the 100kGP. Specifically, we asked participants about their experiences thus far; how they came to be referred to 100kGP and why they took part; what their hopes and concerns were; what they thought about its research aspect, including what they thought about consent to broad research and about their genomic data being made available to researchers; when—if at all--they expected a diagnosis; and whether and how they had talked to family about 100kGP as well as whether their family members were participating. Interviewees gave consent before their interviews, which lasted around 60 minutes and were digitally recorded and transcribed. SD interviewed all participants in their homes or in her research office—participants chose whatever was most convenient for them. We ceased recruitment once we approached saturation (defined as occurring when the main categories have depth and variation (Corbin & Strauss, 2008)) — in this case, after 16 interviews with 20 interviewees (four interviews with couples and twelve with individuals). Most were the parents of patients—just four were patients themselves. Ten had a suspected, but not genetically confirmed diagnosis. Among the remaining participants, there was no diagnosis suspected. To maintain anonymity, we have not disclosed here the rare diseases that affected participants.

We analysed data inductively drawing on grounded theory (Corbin & Strauss, 2014). Co-authors, who have a track record of working closely together on data analysis, analysed parts of the data to improve rigour (Lincoln and Guba, 1985). Specifically, SD took the lead role and AF and AL independently analysed sections of different transcripts; during the analysis period they met regularly to discuss emerging themes and cross-sectional differences and similarities. This process was iterative enabling initial findings to influence the topics discussed during the on-going interviews Interviewees had all been engaged with healthcare for many years to manage the signs and symptoms of their, or their child’s, conditions. Most had been on a so-called ‘diagnostic odyssey’ (Turnball et al., 2018), seeking but not receiving a genetic diagnosis. None had been involved in Genomics England’s participant panel or public and patient involvement (PPI) network. Our two main themes illustrate: (1) interviewees participated because they trusted, cherished, and relied on their healthcare providers, and wanted to improve the NHS; and (2) interviewees had concerns for the future of healthcare: these focussed around the changing nature of their relationships with NHS healthcare professionals, as well as big pharma involvement and big data approaches.

**RESULTS**

**Theme 1: Taking part because of trust in, reliance on, and investment in, healthcare**

Theme 1a: Interviewees participated based on personal trust in the healthcare professional who referred them

Many reported finding aspects of the 100kGP difficult to understand: few had read all the 100kGP information sheets and participants said that they found it difficult to take in the information given during the consent appointment. However, in general, interviewees talked positively about the 100kGP with a few referring to their participation as a “privilege” (P13, P11). Most did not feel the need to understand all the information primarily because NHS healthcare professionals (HCPs) had referred them to 100kGP:

P12: If I didn’t fully understand, it wasn’t a case of go[ing] off and read[ing] this and understand[ing] it, it was just more like I’m trusting, I’m handing her over to the experts.

Participants portrayed healthcare professionals as simultaneously expert and as caring, looking after their interests:

P8: [Dr] got a Nobel Prize for his genome work. He was also well aware I was interested in how genetics has progressed today, and maybe that influenced him in putting me forward for the genome project.

P3: I trust [my consultant] very much, because he’s been so good. [Genetics is] what he knows more about than I’ll ever know. But he’s also very caring in treating each patient he’s got individually, because they are rare disorders. People love to go and see him.

Further, they accepted that taking part in the 100kGP involved some vulnerability, such as loss of control and choice, and this was regardless of how informed their consent was:

P19: With anything, once you've given some kind of consent, you can’t always control what happens to it. There is a bit of a limit to your consent. And where it goes from there, you’ve got to let it be.

Trust, in this context, meant a degree of willingness to enter into a position of vulnerability based on a positive expectation about the behaviour of another individual (Colquitt and Rodell, 2011; Fotaki, 2017) and, in this case, one or more HCPs. We do not mean to suggest that interviewees took part because their feelings towards their HCPs clouded ‘rational’ judgement (Johnsson et al., 2013), or because of any ‘therapeutic misconception’—i.e. thinking the 100kGP would benefit them in ways that it would not (Hallowell et al., 2009). Indeed, as we explore next within this theme, most people’s expectations about the likelihood of getting a diagnosis, and about the clinical utility of any diagnosis, were realistic. For this reason, they placed more emphasis on receiving good ongoing frontline care; for example: having good relationships with HCPs, and access to prompt appointments and high-quality treatments, than on getting a diagnosis.

Theme 1b: While a diagnosis might be welcomed, good frontline care is crucial

Unsurprisingly, to some extent, interviewees did want a diagnosis for their or their child’s condition, and for many, the potential for WGS to provide this was the reason for their participation. Nevertheless, many thought that a diagnosis would be unlikely and perceived only minor immediate benefits of receiving one, such as applying for “travel insurance” (P15) or satisfying “medical interest” about the underlying cause of the condition (P12; P4). Low expectations around getting a diagnosis arose from earlier experiences with genetic studies (P18) which had left them aware of how complex it was to reach one; and because the person seeking their consent had told them that a diagnosis was not guaranteed. As illustrated by P3, whose doctor had previously misdiagnosed him, the lack of diagnosis and non-guarantee of a diagnosis from the 100kGP, had not led to any loss of trust in HCPs, it had simply shaped their expectations:

P3: To be told, ‘I’m sorry, you were misdiagnosed eight years ago’. It’s incredibly brave of him to do that. He’s a lovely guy and he’s very, very caring in lots of ways.

This supports the notion that openness can enhance trust and that it grows over time (Fotaki, 2017); this is especially pertinent because most interviewees had been engaged with the healthcare system for many years. Interviewees were also aware that, even if they did receive a diagnosis, it would unlikely supply answers about a prognosis, or lead to a “cure” or any more effective treatment:

P17: Even if there is a [diagnosis of a] rare condition, are we going to have those answers of what’s going to happen tomorrow? Probably not. I don’t mean to be pessimistic about it; I’m just being realistic. We just don’t know.

P12: A cure is very unlikely. It would be a bit of a miracle, really.

Thus although a diagnosis would be welcome many were realistic about its likelihood and/or its impact. Moreover, participants were often very focussed on clinical care and some were confident that they were already receiving the best treatments available:

P15: It’s not going to change her treatment, because everyone’s doing as much as they can anyway. It’s not going to [lead to] a miracle cure.

Moreover, a few expressed some fears about receiving a diagnosis in case the condition they had been adapting to was the wrong one, or in case it revealed a worse than expected prognosis. Adapting to their own, or their child’s condition, had been difficult, but over time, they had learnt to cope with symptoms and ultimately thought a genetic diagnosis would do little to change their day-to-day living experience:

P12: Time passes and you’re more accepting that this is how your child is. [A diagnosis] becomes less important because you’re just getting through every day. When you have a mentally and physically disabled child need[ing] 24-hour care, you’re caught up with that more than anything.

Experiences of good ongoing care from HCPs, and by extension the NHS, factored hugely and helped them cope with their own or their child’s illness. Participants placed immense value on this care:

P2: They’re brilliant with [patient son]. Oh yes, up the NHS!

P20: The treatment and the medication he’s been on, the NHS, it’s really been brilliant. Top doctors. You couldn’t ask for anything better...the hospital staff, the nurses, everybody was brilliant.

Getting a diagnosis might be viewed as the theoretical ideal outcome of their participation, but generally participants agreed that good clinical care was of the utmost importance. They trusted their HCPs to do their best. Despite not necessarily expecting a diagnosis, interviewees seemed sold on the promise that the 100kGP would improve care for their family and more widely eventually—something they were invested in facilitating, as we discuss next.

Theme 1c: Investing in improving the NHS

Interviewees saw 100kGP as the first of many steps towards some type of “answer” (P13, 52), ones that might not help them personally, but that might benefit their children, grandchildren, society, and healthcare more generally. Interviewees did not perceive the uncertainty around whether 100kGP would help them personally as “a bad thing” seeing their contribution to improving NHS care as important:

P19: Any experiments that are happening in any of those fields, it takes quite a long time for them to be [adopted into care], and not everything is effective for everyone.

P18: It’ll be a step forward. Quite where it goes I don’t know, but it’s going to start giving more data. It’s answers—it might not be the answers we want or were looking for—or it['ll] create more questions. But it’s just information, and that’s not a bad thing.

As illustrated participants did not buy into genetic determinism, or ‘genohype’, i.e., that sequencing would lead to a diagnosis which would lead to a treatment. They understood that they were contributing to a long-term project, believing (or at least hoping) that genomics could improve NHS diagnostics and treatments for “future generations” (P7):

P1: We’re trying to provide this information to help other families in the future to understand a bit more about [son’s possible condition]. It’s more about the long- term…doing the foundation work to then be able to do research in the future.

P18: Potentially in the future they may be able to switch off genes. If that’s an option that could be available to my family, that would be amazing.

The idea that their contribution could help others made them feel positive; and a few reported that their participation was an act of solidarity—helping others like them:

P16: Being in the special needs world, I know how much it affects people. To be part of something that could help somebody in future generations is a nice thing to do.

A few also talked about long term economic benefit to the NHS; for example:

P20: A lot of the money they’re putting in [to 100kGP] will come back. If you could eliminate, say Huntington’s disease or Alzheimer’s, you’ve saved all that money you’re spending on it at the moment.

P19: If it means avoiding that or diabetes, or something like that, it can save the NHS millions in the future.

Participants understood that the 100KGP was a long term project, and that it came with a certain amount of uncertainty; nonetheless, they were generally confident that it was a positive endeavour that would benefit society. Despite this, interviewees’ discussions raised issues about the future, namely the impact on their relationships with healthcare professionals, and the ethical implications of big data approaches and big pharma’s involvement which we explore in the next theme.

**Theme 2: Concerns about the future**

Theme 2a: Care pathway being interrupted by 100kGP research process

As we have outlined above, participants were understandably focussed on on-going clinical care for themselves and their family and trusted their HCPs and the NHS. However, a few had less positive experiences of the research nurses/clinical trials assistants who led the consent process for their involvement in 100kGP—staff they met for the first time during this process and who some felt were less experienced and less expert in their particular condition:

P8: It would be nice to have somebody who was pretty knowledgeable. I don’t want to knock the two [research nurses] doing the consent forms, but they were struggling.

Participants’ previous genetic tests had been integral to their clinical care, but now suddenly they had been given a separate appointment with someone they had not met before to give consent for whole genome sequencing and few remembered the name or professional role of this person. Some commented on staff members seeming intimidated by, and inexperienced at dealing with, the family:

P18: Because there was a group of us, it, I think it was a bit daunting for the gentleman unfortunately.

And this had the potential to impact negatively on the consent process:

P1: The research nurse wasn’t particularly au fait with children, so [her] taking them off and giving us the time to then go through [the consent process] wasn’t really an option. So yeah there wasn’t really that space to ask the questions.

Given the new team of staff involved and its detailed nature, the consent process for the 100kGP appeared disembodied from the care they had received thus far. Interviewees also felt confused about who would conduct follow up appointments with them: when, how, and from whom they would receive any results; P17 was frustrated at the lack of clarity on this in Genomics England’s patient-facing information:

P17: I don’t know whether it would be [the person who sought consent] that would give the results or whether it would go straight to the neurologist, who initiated the referral onto 100,000 genomes...There doesn’t seem to be any clarification on who is giving what information out, when there is information to give out. There’s no definitive person.

Having had at least two lengthy discussions with staff members, as well as reams of information, in the run up to their consent appointment, the sudden silence that followed the appointment disquieted some. So, while on one hand the uncertainty around whether they would get a (helpful) diagnosis was one for which they had a fairly high tolerance, interviewees were less tolerant of uncertainty about their future care pathway. Others echoed this sentiment highlighting how the disjunction between the clinical and research functions of the 100kGP had the potential to impact on clinical care:

P3: Do they ring [son] up? Do they write a letter? I don’t know. We don’t know. None of us know. It's not so good, because you’re willing to take part, but it would be nice if you got some contact. We’re just in limbo at the moment.

P13: Do they contact you? I don’t hear anything, what does that mean? Am I going to hear something if it’s nothing at all? How will I be communicated to about anything?

Comments such as P3’s “you’re willing to take part, but…” hints at an expected reciprocity from Genomics England – or their delegates. Having given up their time freely, they expected better communication than they had thus far encountered. They also worried that non-expert professionals might end up giving them their results, and that they would have to negotiate their own way through future clinical care:

P17: There’s nothing worse than being told something and it’s like, 'well I don’t actually know the answer to that, I’d need to make another appointment'.

Interviewees reported trust in their ongoing relationships with their HCPs and Kelly et al. (2015) have previously shown the importance of an ongoing relationship between interviewees and researchers for establishing trust. However, the 100kGP (as well as any other research studies they had taken part in) had introduced a range of new actors: staff seeking consent, Genomics England, and different HCPs whom they might not have seen previously. The 100kGP confused their expectations around their relationships with these actors, sometimes creating anxieties about future care.

**Theme 2b: Rationalising big data and big pharma: threats or opportunities for the NHS?**

As well as these worries about their care pathway, several interviewees raised concerns about contributing to research to which they had a moral objection (e.g., P20 “military research”, P1 “cloning”, P8 “‘designer babies”, and research by pharmaceutical companies). Affirming an ethical issue that Genomics England and the Generation Genome report both recognise, interviewees also worried about data security and privacy, and in turn, possible risks of employment/insurance discrimination against themselves and their family. As already highlighted, interviewees recognised that participation might involve giving up some level of control. As such, they rationalised their decisions to take part anyway: first, by saying their desire for potential ‘answers’ and for the future benefit of the NHS and others, outweighed their concerns:

P18: I don’t agree with the costs [pharmaceutical companies] pass back to people [and] the NHS. They make money out of my blood! A lot of money. You see on the news what they charge for producing these things.

I: But for you it’s worth doing because..?

P18: Answers. Treatment. Less painful treatment. Prevention even. Possible annihilation of it all; so that would be wonderful.

Secondly, they perceived it as a ‘normalised’ aspect of society for government bodies to amass personal data and for pharmaceutical companies to be involved in the NHS. They argued that declining participation would do little or nothing to change these aspects:

P4: Your fingerprints are on a database... the police took my fingerprints... I imagine somewhere you’ve got records everywhere.

P6: About drugs being manufactured and possibly people making money out of things, that doesn’t have any bearing on it at all to me. It’s something that’s going to happen regardless isn’t it … It’s just the reality of the world, it’s the way it works isn’t it. No-one does anything for nothing.

Thirdly, they felt reassured about data security because they trusted the institutions involved - NHS hospitals, Universities, and ethics oversight committees. Although no interviewee indicated a detailed knowledge of the oversight involved, speaking instead in vague terms about the mechanisms in place, they used language showing a sense of certainty that the involvement of this oversight would protect the biorepository and their data:

P1: The fact that ethics will have to be involved, my understanding of ethics alleviates the concern. There’s such strict regulation over ethics now, that it will only possibly be used in the right way. [emphasis added]

P16: It’s done through the University and hospital, and it will be carried out professionally and with all the relevant confidentiality things in place. [emphasis added]

Some interviewees did express concern about oversight in the slightly more distant future, questioning whether data security, privacy, and access regulations would be as ‘strict’ in years to come, hinting at the need for funding of infrastructure and experts to continue post-100kGP. One interviewee, with a computer science background, thought data in the biorepository ought to remain nationalised, that it should be licensed (rather than sold or given away) to, for example, pharmaceutical companies, and that these companies should not be able to patent drugs developed using NHS data:

P20: It should be an open source. If someone wants to do a project, they should be allowed to, and not have restrictions put on [e.g.] a pharmaceutical company controlling it, saying they’ll do [the research] and...patent…drugs. That only slows things down....Maybe [pharmaceutical companies] ought to have a licence for developing the drugs for ten years or whatever, and they shouldn’t be able to patent anything.

This interviewee thought that any beneficial development created using NHS data should flow back to the NHS because the national-ness of the health service has made the 100kGP possible in the first place. Indeed, P8, who also had a science background, hinted at this too, “The NHS is a wonderful, world-class database, which is totally underutilised…so I’m all in favour of it being used”. These interviewees’ occupational backgrounds perhaps made them more attuned to the nuanced ethical issues around big data and big pharma than other interviewees. Generally, however, participants trusted institutions such as the NHS and research governance procedures to keep their data secure and use it for the benefit of society.

**DISCUSSION**

This paper has interrogated the views and experiences of patients, and family members thereof, participating in the 100kGP. Our first subtheme highlighted the key messages of this paper: people’s decisions to participate in 100kGP were based on trust in the HCPs and the NHS; and that the NHS was an institution that they relied upon and were invested in improving. In effect participants trusted in the 100kGP by proxy, as they saw 100kGP as being an extension of the NHS and clinical care. These findings support research from Genetic Alliance, which found that most patients, and family members, welcome research initiatives with the NHS at its heart, as well as supporting Busby’s (2006) research about why patients (recruited through the NHS) participated in genetic research. Like us, Busby found that the detail of the study was of little concern; that they thought research would help future generations; and that their involvement was based on a direct “investment in[to] the future health of an imagined community, in which the NHS featured prominently” (p214). Our findings also reflect previous research showing that decision-making about whether to take part in a research study or a biorepository is not based solely on a rational choice following a weighing up of harms and benefits, but also on the complex interaction between personal, cultural, social and economic circumstances, and in the institutional context where consent is sought (Hallowell et al. 2009; Martin and Hollin, 2014; Samuel et al., 2017; Timmons and Vezyridis, 2017).

Genomics England reiterated that the 100kGP should be “by the NHS, for the NHS”, (Martin and Hollin, 2014, p14). Indeed, Woods (2016) pointed out that Genomics England rallied the public to a common cause and called upon the public’s ‘civic duty’ to endorse and participate in 100kGP and arguably, so too did the Chief Medical Officer when saying: “*to make this [genomic medicine service] dream a reality...we need to...agree to use of data for our own benefit and other.*” (Ch1, p. 4). This hints at the moving boundary between research and clinical practice in genomics, and our data highlights some of the potential issues this raises. A diagnosis in genomics will often only be possible if data is compared with others and it is this aspect that makes it different from other types of healthcare. It should be noted that interviewees were not completely ‘altruistic subjects’—ideally, they would have liked a diagnosis. Nevertheless, their desire for one was neither universally nor strongly articulated. They had a high tolerance for uncertainty in this respect, being accustomed to not knowing what the genetic diagnosis was. While interviewees did not appear to believe in genomic determinism, they were hopeful in relation to the promissory aspects of the genomic venture, i.e. that it could make the NHS better. Indeed, improving the NHS was important to interviewees—not least because it was a form of making some meaning from their difficult experiences.

Because they were so reliant on the NHS, interviewees were somewhat less tolerant of (i.e. more anxious about) uncertainty regarding their future care pathways, such as who would notify them about the outcome of their sequencing analysis and whether efficient follow up care would be available. These worries came alongside a feeling that staff they had seen as part of their consent conversation for the 100kGP were not always seen as expert or experienced. Our theme 2a illustrated the impact of the care pathway being interrupted by the research process of the 100KGP. Participants were happy to take part, but this was based on their experiences from long term engagement in healthcare and the development of trust over time, which might easily be eroded. These findings echo Day et al.’s ethnography of a cancer service, which offers a warning call for how genomic and precision or stratified medicine could affect patients’ experiences of relationships with their clinicians. Day et al. (2017, p154) found:

“stratified medicine placed additional strains on the service through its requirement for a highly-skilled workforce and a meticulously integrated patient pathway that, in the context of budget constraints, were difficult to deliver. Highly-skilled staff have moved increasingly to back-office functions such as laboratory analysis [and] replaced in frontline functions by less qualified staff…Some patients describe care that is far from personalised.”

These findings call into question whether the move of skilled staff to back-office functions, and less qualified staff to patient consultations, will become more widespread as the genomic ‘revolution’ rolls out. NHS England is funding a range of courses for the ‘mainstream’ NHS workforce to ‘upskill’ their genomics knowledge, as well as genetic counsellors to train in bioinformatics (NHS Health Education England, 2017). Such training is crucial but even if this is improved, clinicians might have less time with their patients as genome testing increases and some skilled work might be left to mainstream specialists without the degree of experience that those in clinical genetics will have. Financial cutbacks are making this more common (Maynard, 2017) and a recent UK government report has highlighted that there is still insufficient training and a lack of qualified staff to fill posts (House of Commons Science and Technology Committee, 2017).

Other concerns that our interviewees raised about data security, privacy, and access, e.g., by commercial companies, were more in line with the sorts of issues Genomics England and the Generation Genome report have identified as ethically problematic. Our interviewees, much like the 100kGP participants Genomics England have featured in public engagement videos, indeed saw large-scale data (‘big data’) collection and commercial (‘big pharma’) involvement as contentious, but not enough to decline participation. Other studies about biobanks and research also show that members of the public and patients accept that commercial companies will access their data, although this acceptance is sometimes reluctant and caveated by a need for transparency (Nicol et al., 2016). Such survey findings can be limited in validity: first because participants are likely basing their views on an incomplete understanding of how big data/big pharma affects the NHS (ComRes, 2017[[5]](#footnote-5))—especially given the lack of transparency around such issues (Carter, Laurie, and Dixon- Woods, 2015; ComRes, 2015; Powles and Hodon, 2017; Sterckx et al., 2016; Vezyridis and Timmons, 2017). Second, participants’ arguments were not always sound: for example, the normalisation of large-scale data collection and pharmaceutical involvement in the health service does not mean something is ethically acceptable. We would thus argue against taking the findings of studies that ask about such complex issues, but without informing and engaging respondents about the ways data are used, as evidence of ‘trust’ and in turn as reasons to legitimise data-sharing and other plans. Sheikh and Hoeyer (2017) similarly contend:

“glib policy statements about ‘trust’—and how to ‘win’ it, can end up legitimizing uses of people’s donations (following an implicit argument of the type ‘we can use this biomaterial because we would not have it if people did not trust us’) without paying adequate attention to the actual reasons people have for participating in research.”

Although our interviewees trusted (what they knew of) the oversight mechanisms involved in 100kGP, they worried about what oversight there would be in the slightly more distant future. A couple of interviewees, both with a relatively sophisticated understanding of data ethics, importantly thought the biorepository should be a national asset that could benefit the NHS and society. This finding supports a Wellcome Trust/Ipsos MORI study about trust and commercial access to data, which involved in-depth workshops with patients and members of the public, which helped them understand existing data-sharing activity in the public and private sectors, and found that participants:

“[did] not want anyone (public or private sector) to be able to co-opt health data for political ends, for example giving it to organisations who might have an interest in dismantling the NHS. For example, if work that the NHS could do is done instead by private companies who succeed because they have access to public data, this is felt to be wrong.”(Wellcome Trust/Ipsos MORI, 2015, p102)

The recent Life Sciences Strategy report has similarly suggested that the UK should not give away biorepository data but rather charge commercial companies to access it or seek some other form of benefit sharing for the UK tax payer and NHS patients (Bell, 2017; Science and Technology Committee evidence, 2017).

Given all of this, a practical recommendation that comes out of this research is that NHS England should listen to, and take seriously, patients’ and public voices. They could do so by maintaining the current Genomics England Patient and Public Involvement (PPI) groups and Public Engagement events, and ensuring patients are represented on the Access Review Committee, which decides who should have access to genomic data and for what purpose.

**Conclusion**

While this current study presents the views of a sample that is self-selected (in two senses: those participating in 100kGP and willing to be interviewed), from one cohort (the rare disease group), and from one Genomic Medicine Centre (albeit covering population of 3 million) it provides a unique perspective into user’s views about a health service ‘transformative’ project. To date such views have been mainly sought from hypothetical users or members of the public. Many of our participants were early recruits and they – or their children - had therefore perhaps the longest odysseys, while in the future whole genome sequencing will be offered earlier in the care pathway. Our data provides useful insights just as genomics is poised to become NHS business as usual and reveals that, while participants had some concerns about the consent process itself, they had more worries about downstream communication about any sequencing results. We have made our research context explicit so that others can determine how the findings might apply in other similar areas. However, it must be borne in mind that as genomics becomes routine, there will be different patients who might have different views, for example may be more or less trusting in the NHS or more positive about their experiences of communication.

We found that interviewees’ decisions to participate in 100kGP were based on interpersonal and institutional trust in the NHS, and on an investment in improving care for the future. Interviewees relied upon receiving good ongoing NHS care for managing their own or their child’s rare disease, but they worried about what their relationships with NHS healthcare professionals would be like in future. A few participants worried about whether Genomics England’s biorepository would remain protected and an asset of the NHS. To honour and foster participants’ trust, which may easily be lost, and their ‘clinical labour’, we therefore recommend ongoing public engagement and consultation about how genomics is integrated into the NHS (especially given its current funding and staffing constraints) and about what happens to the data as Genomics England hand over responsibility to NHS England.

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Figure 1

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Step 1  Healthcare | Step 2  Genomic | Step 3  At least 24 hours | Step 4  Samples sent to | Step 5  Genome data are |
| professionals | medicine centre | later, staff see | the sequencing | combined with |
| treating | staff contact the | patients (and | hub, and the | patient data from |
| potentially | potential | family members) | sequence data | hospital/clinic/GP |
| eligible patients | participants and | in a face-to-face | are sent for | records, national |
| refer them to | send information | consent | storage to | disease registries, |
| 100kGP | documents | appointment | Genomics | social care |
|  |  | where they take | England’s data | records, and |
|  |  | blood samples. | centre. | Public Health |
|  |  |  |  | England, |
|  |  |  |  | extracted over |
|  |  |  |  | the patient’s |
|  |  |  |  | whole life. |

1. The report uses all three terms. However, they are not synonymous. See Day et al. 2017. [↑](#footnote-ref-1)
2. The list of rare diseases eligible for 100kGP can be found on Genomics England’s website <https://www.genomicsengland.co.uk/information-for-gmc-staff/rare-disease-documents/> [↑](#footnote-ref-2)
3. NHS England is an executive non-departmental public body of the Department of Health. NHS England oversees the budget, planning, delivery and day-to-day operation of the commissioning side of the NHS [↑](#footnote-ref-3)
4. Given the timing of our research, the participants in our research were early recruits to the 100KGP

   and were from the rare disease group as cancer patients were yet to be recruited into the 100kGP [↑](#footnote-ref-4)
5. Their survey with over 2000 people showed that many people (33%) “know very little”(33%) or “nothing” (6%). Percentages are likely to be higher in reality: self-report surveys are vulnerable to people overestimating their knowledge (known as the Dunning-Krueger effect). [↑](#footnote-ref-5)