

# GENOME SEQUENCING AND THE NHS

The views of rare disease  
patients and carers



**GENETIC  
ALLIANCE** UK

**Genomics**  
england



# ABOUT GENETIC ALLIANCE UK



Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 200 patient organisations. We undertake various initiatives to improve health service provision, research and support for families. These initiatives include:



Rare Disease UK, a multi – stakeholder coalition brought together to work with the government to effectively implement the UK Strategy for Rare Diseases.



SWAN UK (syndromes without a name), the only UK-wide network providing information and support to families of children without a diagnosis.

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# INTRODUCTION

In October 2018, Secretary of State for Health and Social Care, Matt Hancock MP, announced the launch of the Genomic Medicine Service.

The service will offer whole genome sequencing (WGS) to patients with certain rare and undiagnosed conditions, and certain cancers, as part of their routine NHS care. In 2019 this will be extended to all seriously ill children, including those with suspected genetic conditions and cancers.

This initiative sees the commitment of the UK government to sequence 5 million genomes over a 5-year period, and firmly places the NHS as one of the leading healthcare systems in the world to use whole genome information in mainstream clinical practice.

## What is whole genome sequencing?

Whole genome sequencing (WGS) is an approach that allows the reading and analysis of a person's entire genetic code.

The introduction of WGS into the NHS holds the promise of earlier diagnosis for rare disease patients, which can be the first step to accessing the most appropriate care, information and support. It will also enable research into diseases and treatments, and planning for healthcare services.

## What is the 100,000 Genomes Project?

The 100,000 Genomes Project, launched in 2012, was established by the Department of Health in

order to carry out the sequencing of 100,000 whole genomes.

Part clinical, part research, the 100,000 Genomes Project sequenced the genomes of patients with specific rare diseases, cancers and infectious diseases. The Genomic Medicine Service will build on the 100,000 Genomes Project with the aim of transforming NHS genetic services by integrating WGS into mainstream clinical practice.

The UK has become the first nation in the world to apply whole genome sequencing at scale in direct healthcare.

## What was the aim of this project?

Introducing WGS into mainstream NHS care raises important questions for patients and carers. Therefore, it is vital that their views, as potential beneficiaries of such a service, are sought in order to help guide decision-makers in policy and healthcare.

In 2014 Genetic Alliance UK conducted a survey, commissioned by and in collaboration with Genomics England, to capture the views of patients and carers to help inform the implementation of the 100,000 Genomes Project. Four years later, as NHS England and Genomics England prepare to deliver the Genomic Medicine Service, it is important that patients and carers are once again consulted.

Genetic Alliance UK were again commissioned by Genomics England to carry out a consultation and this report summarises the findings from this work.

# SUMMARY OF KEY FINDINGS

## Participants' experiences of the 100,000 Genomes Project

1. The majority of our respondents were glad they had taken part in the 100,000 Genomes Project (73%), would take part again (86%), and would be likely to participate in future medical and genomic research.
2. However, fewer than half (43%) of our respondents said they were 'satisfied' or 'very satisfied' with their overall experience of taking part in the 100,000 Genomes Project. Respondents indicated that the benefits they hoped for from taking part were not matched by actual benefits achieved at the time of the survey.
3. Most respondents (77%) had yet to receive a result at the time of our survey, which offers a likely explanation for the discrepancy between findings 1 and 2.
4. Over 80% of patients and carers felt they had been provided with sufficient, comprehensible information about the 100,000 Genomes Project before taking part. However they would have liked more information about what to expect during the process, and more regular contact while waiting for a result.

## Participants' views on the provision of WGS through the NHS

5. The most commonly hoped-for benefits of WGS through the NHS were: improvement of the individual's care or treatment, contributing to research into their condition, and obtaining a diagnosis.
6. Knowledge of how different organisations use health data collected during NHS care is limited amongst patients and carers. Although they are willing for healthcare professionals and researchers in the NHS to have access, around a quarter of respondents 'might decline' WGS because of concerns about wider access to data.
7. Patients and carers recognise the value of using data generated from WGS for research purposes – and the vast majority support it. However they also feel it is important that there is choice about whether their data is used in this way.
8. Patients and carers overwhelmingly believe that, if different organisations will have access to their WGS data for research, this should be with the expectation that there will be a clear public benefit. Respondents identified data misuse and data security as potential risks.

# CONCLUSIONS

The Genomic Medicine Service, which will build on the 100,000 Genomes Project and deliver genomics as an integral part of the NHS, has already been officially launched. In this sense it is timely to review the experiences of rare disease patients and their carers who participated in the 100,000 Genomes Project, and to scope out expectations and concerns for the Genomic Medicine Service.

But the majority of rare disease patients who participated in the 100,000 Genomes Project have yet to receive a result. It is of note that despite this, a large majority of respondents were glad to have taken part in the 100,000 Genomes Project, reinforcing the perception that there is a great deal of support for the application of genomics among rare disease patients and their carers. There is optimism about the ability of the Genomic Medicine Service to deliver improved care and treatment, to contribute to medical research and to provide diagnoses.

The positives from the 100,000 Genomes Project now need to be taken forward into the Genomic Medicine Service, and necessary improvements need to be implemented.

Respondents to our survey were very positive about the information they had received before taking part in the 100,000 Genomes Project, but there are instances of miscommunication about what analysis would take place and how long the process would take. A separate recent study has indicated there is room for improvement in the

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1 - Saskia C. Sanderson PhD, Celine Lewis PhD, Christine Patch PhD, Melissa Hill PhD, Maria Bitner-Glindzicz MBBS, PhD & Lyn S. Chitty PhD (2018) 'Opening the "black box" of informed consent appointments for genome sequencing: a multisite observational study' MRCOG Genetics in Medicine <https://doi.org/10.1038/s41436-018-0310-3>

ability of healthcare professionals to deliver fully informed consent for genome sequencing<sup>1</sup>. As the Genomic Medicine Service develops, protocols and staff training must place an appropriate emphasis on effective communication with patients and carers about genome sequencing.

This communication is in three phases:

- before testing, where information about the purpose of the test, what might be found, what might be done with the results and who the results might be shared with, is delivered;
- before the results, where a dual approach to communication is necessary while individuals await a result: regular updates to patients and carers would reassure that they have not been forgotten, and access to an effective online tracking system will allow patients and carers to access an update when they choose to. These open channels will improve confidence that progress is being made with individuals' samples. Although the Genomic Medicine Service promises a faster turnaround than has been possible through the 100,000 Genomes Project, there is likely to remain a core of patients who face a long wait, for example those who are told that nothing has been found with the initial analysis but for whom further analysis might be promised;
- at the delivery of results, where healthcare professionals delivering results from WGS must be trained in interpreting reports: variants of unknown significance (VUS) that warrant follow-up must be explained, and access to genetic counselling must be available where appropriate.

We should acknowledge that the 100,000 Genomes Project has not yet given us the volume of

experience at the end of the process - in feeding back results - that it has at the beginning in enrolling patients and empowering them to provide informed consent. It is likely that there is more to learn on this aspect of genomic healthcare provision.

Consistent with the emerging picture of patient and public attitudes to access of health and genomic data, our respondents see the value in using genomic data for medical research, but this comes with caveats.

There must be an expected public benefit; individuals may wish to have the power to choose whether and by whom their genomic data is accessed (with fewer respondents being supportive of access by commercial organisations than by the public sector); and data misuse and security are identified as potential concerns. An effective public discourse, and a consistently applied communication process within the NHS,

will help address these issues.

The Genomic Medicine Service promises a cutting-edge approach to diagnosis, so it is appropriate to sound a note of caution about the post-diagnostic service available to patients, and its potential impact on patient satisfaction with health services.

The NHS is resource-constrained and specialised services are updated infrequently at best. In addition, results from WGS might not be clinically significant at the time they are reported back to patients, but could become actionable in the future - and in some cases research will be needed to deliver a meaningful finding. In these cases there must be a clear message to patients and carers as to what to expect, and who has responsibility for renewing contact when appropriate.

It is clear that whether the Genomic Medicine Service ultimately delivers on its promise will depend on factors outside pure progress in genomic technology.

## METHOD

Genetic Alliance UK developed and hosted an online survey (with input from Genomics England) which was launched in October 2018. The survey was sent out to: the patient groups that make up the membership of Genetic Alliance UK; supporters of the Rare Disease UK campaign; and families of children with an undiagnosed genetic condition who are members of SWAN UK. Patients and carers of individuals with rare or undiagnosed genetic conditions were invited to take part.

The survey was live for 27 days and received a total of 597 responses. The survey explored patients' and carers' views in relation to two key areas:

- What was the experience of those patients and carers who have taken part in the 100,000 Genomes Project? (see page 6)
- What are patients' and carers' views about the prospect of WGS being provided through the NHS? (see page 12)

# FINDINGS

## Who responded to the survey?

A total of 597 UK-based individuals responded to the survey. This included patients (54%), parents of patients (56%), other blood relatives (5%) and carers (who are not parents) of patients (3%).

The vast majority of survey respondents were female (87%). Amongst carers, 54% cared for male patients and 42% cared for female patients.

Almost half of respondents (45%) have a diagnosis for their/their relative's condition, and 91% of these have (or care for someone with) a

rare disease. Of the 12% reporting a 'possible' diagnosis, 80% said this was a rare disease. Among our respondents, 24% have (or care for someone with) an undiagnosed genetic condition. The situation for the remainder of respondents was more complex, reporting more than one condition, some of which are undiagnosed.

Respondents proportionately represented all regions of the UK, with the majority from England (85%).

## Participants' experiences of the 100,000 Genomes Project

### Who had taken part?

Around a third of survey respondents had taken part in the 100,000 Genomes Project. Of those that had taken part, the majority had heard about it through a healthcare professional (86%). This was predominantly a healthcare professional working in genetics (56%), although some were recruited by those in other specialties including neurology, paediatrics and immunology. Usually this healthcare professional was involved in the person's routine care (69%).

Other participants (14%) had found out about the 100,000 Genomes Project themselves rather than it being suggested by a healthcare professional.

### Who had not taken part?

Respondents that had not taken part in the 100,000 Genomes Project were asked to indicate the reason for not participating. The most frequent reason was not having heard of the 100,000 Genomes Project (65%), followed by uncertainty regarding eligibility

(23%) and that the 100,000 Genomes Project had stopped recruiting in their region (12%).

Only a small number of respondents (6%) reported they believed they were eligible to take part but that they had decided not to (or were not able to), because, for example, they were involved in another research project, or because they faced specific logistical issues.

### Key finding 1:

**The majority of our respondents were glad they had taken part in the 100,000 Genomes Project (73%), would take part again (86%), and would be likely to participate in future medical and genomic research.**

### Key finding 2:

**However, fewer than half (43%) of our respondents said they were 'satisfied'**

or ‘very satisfied’ with their overall experience of taking part in the 100,000 Genomes Project. Respondents indicated that the benefits they hoped for from taking part were not matched by actual benefits achieved at the time of the survey.

### Key finding 3:

**Most respondents (77%) had not yet received a result at the time of our survey, which offers a likely explanation for the discrepancy between findings 1 and 2.**

When reflecting upon their overall experience, the vast majority of respondents reported that they were glad that they participated in the 100,000 Genomes Project (73%), and that they would choose to participate again if they could go back in time (86%). Most also said that their experience of taking part would encourage them to take part in

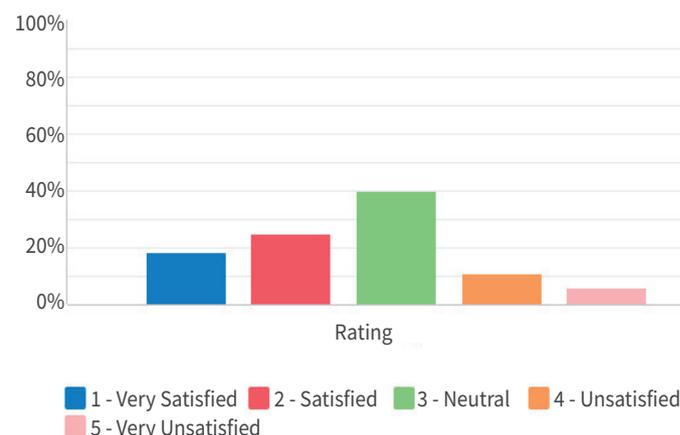
2 - Genetic Alliance UK (2016). The Genomics Conversation  
Genetic Alliance UK: London

**Table 1: Hoped-for benefit compared with actual benefit of the 100,000 Genomes Project**

Benefit	Hoped-for benefit: % of respondents	Actual benefit: % of respondents
Obtain a diagnosis	78%	8%
The knowledge that I am contributing to research into my condition*	70%	35%
Find out if the condition has a genetic cause	70%	7%
Understand the risk to other family members	51%	7%
Improve my own care/treatment*	46%	6%
Predictions about my health*	46%	5%
Receive new information about my health, or potential health problems, not related to my known condition*	42%	3%
The knowledge that I am contributing to research into other medical conditions (not just my own)*	41%	26%
Helped with family planning	22%	6%
Other	2%	16%
The knowledge I have done all I can to try and secure a diagnosis	n/a	35%
I do not feel I have gained anything from taking part	n/a	32%

\*or that of the person I care for

**Chart 1: Respondents’ overall experience of the 100,000 Genomes Project (% respondents)**



further genomic (65%) and medical (56%) research.

Respondents were invited to evaluate their overall experience from 1 (very satisfied) to 5 (very unsatisfied). As shown in chart 1, 43% reported being satisfied or very satisfied, and 40% were neither satisfied or unsatisfied. A smaller proportion (17%) were unsatisfied or very unsatisfied.

The 100,000 Genomes Project raised the prospect of a variety of benefits for patients and families. Some of these benefits may have a direct impact

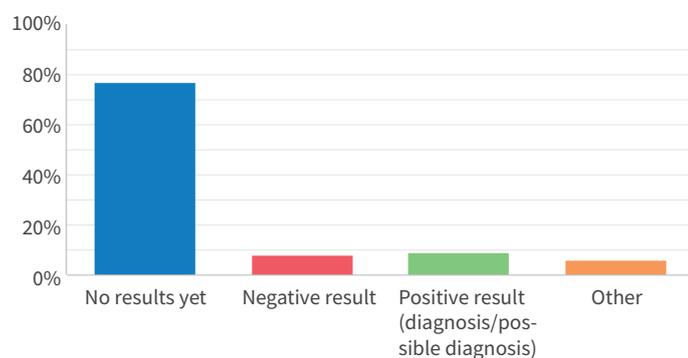
on the individual or family, and some may have a wider impact on society (but not necessarily a direct benefit to the individual).

Survey respondents were given a list of possible benefits that could be gained from WGS, compiled from previous research<sup>2</sup>, and asked to indicate which they had hoped for when they joined the 100,000 Genomes Project. Respondents were then asked to indicate which of these they had actually gained from taking part. As shown in table 1 (page 7), respondents reporting actual benefits were far fewer than those reporting hoped-for benefits.

At the time of the survey most participants (77%) had not yet received a result from the 100,000 Genomes Project.

Only 17% reported receiving a result: 9% (15 individuals) had received a positive result, and 8% (13 individuals) had received a negative result (ie nothing found with the analysis done so far). The data is shown in chart 2. More information about the experiences of individuals who had received a result is presented on pages 10 and 11.

**Chart 2: Results received through the 100,000 Genomes Project (% respondents)**



When invited to indicate whether the 100,000 Genomes Project had met their expectations only 29% of respondents agreed or strongly agreed that it had. This is unsurprising given that most respondents had yet to receive a result through the 100,000 Genomes Project.

Individuals who reported not having received a result yet were significantly more likely to report not having gained anything from taking part in the 100,000 Genomes Project.

However, these respondents were also significantly more likely to report gaining the knowledge that they have done all they can to secure a diagnosis,

as a benefit of taking part. They were also significantly more likely to report having gained other unspecified benefits.

### Key finding 4:

**Over 80% of patients and carers felt they had been provided with sufficient, comprehensible information about the 100,000 Genomes Project before taking part. However they would have liked more information about what to expect during the process, and more regular contact while waiting for a result.**

Respondents rated the information they received about the 100,000 Genomes Project favourably (agree/strongly agree): 85% felt they were given all of the information they needed before consenting to take part, and 82% reported that this information had been easy to understand.

Despite the majority of respondents rating the information they had received favourably, better information was one of the most common suggestions respondents proposed when asked how their experience could have been improved. Specifically, respondents alluded to wanting better information about the process of taking part. For instance, several individuals made reference to being given inaccurate timeframes for receiving results. Respondents' experiences demonstrated that there is considerable scope for improvement in how the process of taking part is communicated.

**'We were frustratingly promised a timeframe of a year and nothing was communicated to explain there would be delays past that timeframe. [There was] poor communication, resulting in huge upset/frustration.'**  
Carer, negative result

Others suggested they would have liked more accurate information about the technical aspects, such as better information about the analysis that would (and would not) be undertaken. This included an explanation as to how findings would be determined to be clinically relevant ('pathogenic') or not.

**‘[My experience would have been improved by not being given misleading hope, into what was being looked at.]’**

Carer, negative result

**‘[I would have liked] an explanation that they do not look at everything but only the genes that match the symptoms.’**

Carer, no result

These comments from respondents add to our understanding of why so many reported that the 100,000 Genomes Project did not meet expectations (see table 1). It is clearly important to be provided with realistic information around the process of WGS, including the limitations of the analysis planned for each individual. Expectations need to be sensitively managed and participants should be prepared for the possibility of a negative result.

All respondents (including those that had received results, and those still waiting) were asked to rate the contact they had received throughout their participation in the 100,000 Genomes Project, from 1 (too much contact) to 5 (not enough contact). The majority (76%) of respondents indicated they had received insufficient contact (rating of 4 or 5).

Additionally, those who had not yet received a result (the majority of respondents) were asked to indicate their preference for receiving updates while waiting. Only 18% of respondents still waiting for a result reported that they would only wish to be contacted if the result would have a direct impact on their care. The majority (82%) indicated they would like to receive updates about ongoing research even when it did not impact their care. Of these respondents, 59% wanted information relating to their condition only, whilst 41% wanted this information along with updates about research into other conditions.

Respondents proposed that more frequent and tailored contact, and easy access to progress

updates, during the wait for a result would have improved their experience of taking part. Several respondents expressed the worry and frustration at the lack of contact they had received, sometimes leaving them questioning whether analysis was taking place, or if samples had been received. Others felt more frequent contact would have made them feel more involved in the research.

**‘I didn’t even know whether our samples had arrived and repeatedly had to email before I got an answer just to find out whether we were actually in the system. My experience would have been improved if newsletters and sample tracking had begun at the beginning.’**

Carer, positive result

**‘[I would have liked] better communication, more updates – not person-specific but general, so people feel like they’re involved! It’s people’s lives.’**

Patient, positive result

One respondent highlighted the impact of being sent surveys in headed envelopes while still waiting for a result:

**‘The timescale (25 months) to diagnosis was too long and being sent surveys in headed envelopes before the diagnosis was cruel as I worried every time it was a result and then found myself bitterly disappointed.’**

Carer, positive result

Respondents also indicated that they felt contact could have been made easier. Several described ‘chasing’ Genomics England for progress updates – which could be complicated by the fact it was not always clear who they needed to contact.

Several individuals specifically mentioned the ‘Track My Sample’ system offered by Genomics England. For some this had been helpful. Others found it was not always accurate, and a number of people had faced difficulties accessing it or had not been made aware of it.

## Insight: the experience of patients and carers who received a positive result through the 100,000 Genomes Project

Fifteen respondents to our survey had received a diagnosis or a possible diagnosis, or an uncertain result (a 'variant of unknown significance') through the 100,000 Genomes Project.

The majority had first discussed the result with a healthcare professional involved in their routine care, and this was usually someone from the specialty that referred them onto the 100,000 Genomes Project. For most, this was a geneticist or genetic counsellor. Several respondents had since discussed the result with other healthcare professionals involved in their care, including specialists in genetics, respiratory medicine, metabolic medicine, cardiology, paediatrics, and GPs.

When evaluating their experience of receiving a result, most respondents reported (agree/strongly agree):

- they were satisfied with the way in which they were originally notified that a result had been found (9/14 individuals);
- discussions about the result and its potential implications were handled sensitively by the healthcare professional (10/14 individuals);
- information about the result was effectively shared between relevant healthcare professionals (10/14 individuals).

Half of respondents reported (agree/strongly agree):

- they did not have to wait too long between being notified of a result and meeting with the relevant healthcare professional to discuss the result (7/14 individuals);
- the result and its potential implications were clearly communicated by the healthcare professional (7/14 individuals);
- they were satisfied with information about the condition/result given by the healthcare professional (7/14 individuals);
- they were satisfied with information about additional relevant support (e.g. patient organisations/support groups) provided by healthcare professionals following the result (7/14 individuals);
- they were satisfied with the ongoing support/follow-up support provided by healthcare professionals, following the result (7/14 individuals).

Although the numbers are too small to draw firm conclusions, these findings indicate that there may be room for improvement in the way WGS findings, their implications and appropriate follow-up, are handled.

## Insight: the experience of patients and carers who received a negative result through the 100,000 Genomes Project

Thirteen respondents had received a 'negative result' (i.e. nothing relevant to their condition had been found with the analysis that had been undertaken through the 100,000 Genomes Project).

When evaluating their experience of receiving the result, most respondents reported (agree/strongly agree):

- that it was made clear that their data was still a valuable resource for future medical research (9/13 individuals).

Fewer than half of respondents reported (agree/strongly agree):

- that discussions about the outcome were handled sensitively by healthcare professionals (6/13 individuals);
- that the possible reasons for the negative result were clearly communicated (3/13 individuals);
- that they were satisfied with the way they were originally notified of the result (4/13 individuals).

(The majority had been notified by letter (7) or phone (4), but these numbers are too small for us to determine whether the notification method influenced satisfaction rates)

Although there were too few respondents with a negative result to draw firm conclusions these findings suggest there is room for improvement in the way healthcare professionals communicate with patients and families about negative results.

## Participants' views on the provision of WGS through the NHS

This section of the survey was completed by people who had taken part in the 100,000 Genomes Project, and those who had not.

### Key finding 5:

**The most commonly hoped-for benefits of WGS through the NHS were: improvement of the individual's care or treatment, contributing to research into their condition, and obtaining a diagnosis.**

Survey respondents were presented with a list of potential benefits of WGS through the NHS (consistent with those presented to respondents reflecting on the 100,000 Genomes Project, earlier in the survey), and asked to indicate which they would hope to gain if they were to consent to WGS as part of their NHS care.

The five most popular benefits were: improving their own care/treatment (92% of respondents chose this); contributing to research into the condition (89%); obtaining a diagnosis (82%); finding out if the condition has a genetic cause

(80%); and better understanding the risk to other family members (73%).

Hoped-for benefits from the provision of WGS through the NHS are shown in table 2 (second column). This data combines responses from those who had participated in the 100,000 Genomes Project and those who had not. Individuals who had participated in the 100,000 Genomes Project were also asked what benefits they had hoped for specifically from the 100,000 Genomes Project. These separate responses are also listed in table 2 (third column) for comparison.

Obtaining a diagnosis was an outcome hoped for by similar numbers of respondents both from WGS being provided on the NHS, and from the 100,000 Genomes Project. Other hoped-for benefits were chosen more often by respondents when they were asked to think about WGS through the NHS, in comparison with the portion of respondents who had been asked separately about their participation in the 100,000 Genomes Project. In some cases this was very marked, for example 92% of respondents chose 'improvement to care/treatment' as a hoped-for benefit from WGS through the NHS, compared with only 46% of

**Table 2: Hoped-for benefit of WGS through the NHS compared to hoped-for benefit of the 100,000 Genomes Project**

Hoped-for benefit	From WGS through the NHS: % respondents	From the 100,000 Genomes Project: % respondents
To improve my own care/treatment*	92%	46%
To contribute to research into my condition*	89%	70%
To obtain a diagnosis	82%	78%
To find out if the condition has a genetic cause	80%	70%
To understand the risk to other family members	73%	51%
Predictions about my health*	69%	46%
To contribute to research into other medical conditions	67%	41%
To receive new information about my health, or potential health problems, not related to my known condition*	64%	42%
To help with family planning	39%	22%
For a financial reward	5%	n/a
Other	1%	2%

\*or that of the person I care for

respondents reflecting on the benefits they had hoped for from the 100,000 Genomes Project.

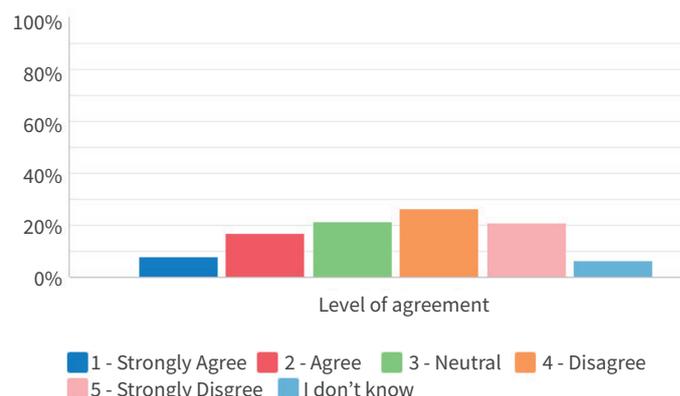
It is perhaps not surprising that individuals who have been selected as eligible, and have gone through the process of informed consent, for the 100,000 Genomes Project have what could be interpreted as more realistic expectations of its outcomes. Although, a deeper analysis of the characteristics of this group of respondents, compared to the rest of the respondents, would be needed to fully understand this finding. Nevertheless it is important that individuals being offered WGS as part of their routine NHS care are fully informed about the likely outcomes so that their expectations are managed appropriately.

### Key finding 6:

**Knowledge of how different organisations use health data collected during NHS care is limited. Although there is support for healthcare professionals and researchers in the NHS to have access, around a quarter of respondents ‘might decline’ WGS because of concerns about access to data.**

When patients use health services, their data is made available to different individuals/approved organisations, for research and planning purposes, unless they have chosen to opt out. In the same way, when WGS is provided through the NHS, the WGS data could also be made available and used in this way.

**Chart 3: Would patients and carers decline WGS if different organisations would be able to access their data? (% respondents)**



Most survey respondents reported having little or no understanding of how different organisations use their health data. This included use by researchers and planners within the NHS (44% said they have little or no understanding), use by external researchers (53%) and - least well understood of all - use by commercial organisations (75%).

Despite the limited understanding of how organisations use data the vast majority of respondents agreed or strongly agreed that they would be willing for healthcare professionals, and researchers and planners in the public sector, to have access to both their health data and their WGS data (see table 3). However, fewer than half were willing for commercial organisations to have access.

Consistent with the findings shown in table 3, 25% of respondents indicated (agree/strongly agree) they ‘might decline’ WGS if their data would be

**Table 3: Who would patients and carers be willing to allow access to their data?**

Who could access data? (% respondents agree/strongly agree)				
	My health data	Health data of the person I care for*	My WGS data	WGS data of the person I care for*
Healthcare professionals	93%	96%	94%	95%
Researchers /planners (NHS)	85%	90%	88%	91%
Researchers /planners (outside NHS)	67%	72%	70%	71%
Commercial organisations	37%	41%	41%	40%

\*answered by carers only

accessed by different organisations in the way described (see chart , page 13). Similarly, 23% of carers said they ‘might decline’ WGS on behalf of the person they care for if their data would be used in this way.

This finding is consistent with previous work by Ipsos MORI with the general public<sup>3</sup> and warrants further consideration. It may be that presenting people with a small amount of information about access to data can trigger concerns, but that these concerns can be reduced when people are provided with the opportunity to ask questions and become better informed.

Our survey provided a small amount of information but no opportunity for further discussion: a good process of informed consent, and a wider public conversation about the purpose of access to health data (and how access is governed), will be important to facilitate a high level of uptake of WGS through the NHS. A key report published in April 2019 demonstrated that there is a willingness to trade complete confidentiality for potential benefits from genomics research, as long as certain red lines around data sharing and data use are respected<sup>4</sup>.

## Key finding 7:

**Patients and carers recognise the value of using data generated from WGS for research purposes – and the vast majority support it. However, many also feel it is important that there is choice about whether their data is used in this way.**

The majority of patients and carers felt that if WGS data is provided through the NHS, it should be made available for research. In fact, 77% felt this data should *automatically* be used in this way.

But this must be placed within the context of the finding outlined above, that respondents clearly have reservations about data access by those outside the public sector.

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3 - Ipsos MORI (2015). The One-Way Mirror: Public attitudes to commercial access to health data. Ipsos MORI:London

4 - Ipsos MORI (2019). A public dialogue on genomic medicine: time for a new social contract? Ipsos MORI:London

Respondents also qualified their support for use of WGS data for research by expressing that this should be properly explained to individuals, and that there should be some kind of consent process.

Respondents were asked for more information behind their answers.

## Reasons for supporting research use of WGS data included:

Benefitting research and understanding, and improving treatment and diagnosis;

**‘As a patient with a rare disorder I understand the need to be able to access genetic data from as many patients as possible in order to try to find genetic causes...to help in future diagnosis and possibly treatments.’**

Patient

Duty to the NHS;

**‘If the NHS is paying for the WGS, then they should be able to use the data.’**

Patient

Helping others, including future generations;

**‘I believe anything that can help not only us but other people and families in the present/future should be done; we know the stress and worry being undiagnosed with rare conditions can cause and would like to think if our participation helped one person not go through that then it was worth it, even if we got nothing from it ourselves.’**

Carer

## Reasons for not supporting research use of WGS data included:

Misuse of data/worries about safety;

**‘There are so many problems with this. Research is done by companies. Who is keeping the data safe and encoding it? They have deals with other companies either in their own stable or elsewhere and data can be shared without your permission.’**

Patient and carer

Sensitivity of data;

**‘I would have concerns about the safety of that data as anonymity cannot be guaranteed...’**

## **insurance companies may use it to decline cover in the future or to better predict inherited risk.'**

Carer

Some respondents identified the concept of balancing possible risks and benefits in order to reach a decision around permitting access;

## **'The chances of being identified are very slim, the benefits of progressing much needed research into diseases far outweigh any risks.'**

Patient and carer

A further important theme was the importance of choice, e.g. the option to opt in or out, or the need for specific consent, for data to be used in this way. Even individuals who indicated they would allow their own data to be used discussed the importance of this being a choice, and that services should not be denied to those that do not agree.

## **'There is so much to learn; the more people we can look at, the faster we will make real progress in diagnosing and treating rare conditions. BUT patients must be willing to give permission for their information to be shared in this way, and those who do not want to give their consent for their WGS info to be shared for research purposes should not be forced to do so.'**

Carer

## **Key finding 8:**

**Patients and carers overwhelmingly believe that, if different organisations will have access to their WGS data for research, it should be with the expectation that there will be a clear public benefit. Respondents identified data misuse and data security as potential risks.**

We presented survey respondents with a list of 11 statements relating to the use of WGS data for research, and asked them to rank their top five most important statements.

The statement 'there will be a clear public benefit' was overwhelmingly considered the most important factor amongst respondents: 74% ranked it in their top five, and of these respondents, 68% ranked it number one.

The full list of statements below is in order of the overall ranking. Note some statements are highly ranked because just a few people chose them but ranked them highly, while other statements were important to more people but they were ranked lower down among their top five.

1. There will be a clear benefit to patients/the public
2. Names and addresses would be removed
3. Data would be securely stored
4. Researchers outside the NHS will have access to the WGS data
5. There will be private commercial benefit
6. WGS data would not be sold on to organisations other than those allowed access
7. Insurance companies would not have access
8. Access to WGS data would be regulated/monitored by an independent body
9. There would be sanctions in place for anyone misusing the data
10. Marketing companies would not have access
11. Private commercial companies will have access for medical research

The list is consistent with other studies<sup>3</sup> that have shown patients and carers recognise the value of research and data sharing. They indicate that most patients and carers support data access, as long as possible risks are controlled, for example by monitoring and protection of sensitive data, and that there is clear public benefit expected of the data access.

## **'If we are not collecting this data for...improving health outcomes through better research, i.e. for the collective good, then it seems of less value and importance.'**

Patient

We also presented a list of possible concerns relating to the use of WGS data for research, developed from our previous research<sup>5</sup>, and asked survey respondents to say which they shared. Again, concerns about data security and access were a priority:

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3 - Ipsos MORI (2015). The One-Way Mirror: Public attitudes to commercial access to health data. Ipsos MORI: London

5 - Genetic Alliance UK (2016). The Genomics Conversation Genetic Alliance UK: London

- Misuse of data (57% of respondents shared this concern)
- Concerns about keeping data safe (52%)
- Concerns about which organisations are allowed to use the data (51%)
- Concerns about privacy and anonymity (45%)
- The potential misuse of genomic technology (40%)
- Whether the NHS has the time/organisational ability (31%)
- Whether NHS staff have the necessary expertise or training (31%)
- Whether this should be a priority for NHS funding (14%)
- Limited effectiveness of genomic medicine (7%)
- Other (1%)

A quarter of respondents said they had no concerns about WGS being provided through the NHS.

Respondents were also asked to consider five different types of data that could be considered

sensitive, and rank them in order of sensitivity from one (most sensitive) to five (least sensitive).

Interestingly, as shown in chart 4, the majority of respondents (51%) felt that their medical records are the most sensitive of the data presented, but only 17% rated their genetic data as the most sensitive. This finding may reflect the fact that our respondents are individuals with direct experience of rare and undiagnosed diseases, who are particularly keen to see benefits coming out from genomic technology. Note though, that there is less consensus among our respondents about the sensitivity of genetic data (the chart shows a more ‘even spread’ of responses), particularly in comparison with medical records which are seen more consistently as the most sensitive type of data.

Despite recent concern over social media companies’ handling of personal data, only 3% of respondents rated their photos and statements shared on social media as the most sensitive category of data. The same percentage (3%) ranked their website browser history as the most sensitive in the list.

**Chart 4: What types of data do respondents think is the most sensitive? (% respondents)**

