

Independent Evaluation of The Generation Study

Views of support organisations



About Genetic Alliance UK

Genetic Alliance UK is an alliance of over 220 charities and support groups working together to improve the lives of people in the UK with genetic, rare and undiagnosed conditions.

Genetic Alliance UK supports people with genetic, rare and undiagnosed conditions by advocating for improved care, progressing medical research, increasing awareness and improving information and support.

We run two long standing projects:



Rare Disease UK: A campaign focused on making sure the UK Rare Diseases Framework is as successful as possible, and to ensure that people and families living with rare conditions have access to a final diagnosis, coordinated care and specialist care and treatment.



SWAN UK: The only dedicated support community in the UK for families affected by a syndrome without a name – a genetic condition so rare it often remains undiagnosed.

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Foreword

The rapid expansion of genomic medicine in the UK presents a profound opportunity to improve outcomes for people living with rare genetic conditions. As genomic technologies increasingly move from research into routine care and as genomic newborn screening is explored at scale, it is vital that these developments are shaped not only by scientific and clinical expertise, but by the insights and experiences of those living and working closest to rare condition communities. Rare condition support organisations are central to this endeavour.

The Generation Study sits within a period of significant policy momentum for genomics. The NHS 10-Year Health Plan signals a strong commitment to embedding genomics within a modern, preventative health service, while the UK Rare Diseases Framework – extended for 12 months from January 2026 – places patient voice and lived experience at the heart of service design and delivery. Translating the aims into practice requires sustained, meaningful engagement with the support organisations that form the backbone of the rare condition ecosystem.

This report draws on the experiences of rare condition support organisations to explore how these organisations have engaged with the Generation Study to date, how prepared they feel to support families, and what is required to enable them to fulfil their role effectively. It also reflects the diversity of the sector: from larger, more established charities to very small, volunteer-led organisations supporting tiny patient populations, often with limited resources but deep expertise and commitment. This is a structural reality of the population of rare condition support groups that needs to be understood and planned for. This evaluation also illustrates the intense drive for progress held within the community of support organisations for rare conditions. The scale of unmet need facing people living with rare conditions generates an appetite for change, improvement and innovation that we expect to endure. Those working in genomic programmes should regard this as a resource to tap rather than a challenge to manage.

A clear message from this evaluation is that support organisations want to be informed, involved and listened to. At the same time, the report highlights significant variation in levels of awareness, involvement and preparedness. These differences are not a reflection of apathy or disengagement, but of uneven communication and the real constraints faced by organisations working with limited capacity.

For organisations whose conditions are not currently included in the Generation Study, uncertainty about the selection criteria and decision-making processes has wider implications. Transparency, responsiveness and clear routes for dialogue are essential, not only to support engagement with the Generation Study itself, but to underpin confidence in future genomic programmes.

We are pleased to see patient voice and support organisations shown to be a valuable tool to link policy, services and lived experience. These organisations provide information and emotional support to families, help navigate complex systems, and offer invaluable insight into how policy decisions are experienced in practice.

The report documents many positive examples of engagement with Genomics England, alongside a clear appetite for continued dialogue. Support organisations want to be constructive partners in the development of genomic programmes, including future iterations of genomic newborn screening. However, the findings also make clear that expectations placed on the sector must be realistic. Small and micro-organisations cannot absorb additional responsibilities without clarity of roles, adequate communication and investment in organisational capacity.

Nick Meade
CEO, Genetic Alliance UK



Executive summary

The Generation Study Process and Impact Evaluation is an independent evaluation run by a team of researchers from multiple organisations, including rare disease support organisations. The views of various stakeholders about the Generation Study are included in the overall evaluation; this report focusses on the views of support group organisations who support rare, genetic and undiagnosed conditions.

Support organisations think it is important to be kept well informed about the Generation Study through a variety of communication channels such as newsletters and emails. They would like to receive regular updates from Genomics England about how the study is progressing as well as being able to find information on how conditions were selected for inclusion and more details about each stage of the study.

Respondents reported mixed experiences with Genomics England's responsiveness if they had been in touch with them ranging from frustration over unanswered queries to feeling heard. One of the main reasons for support organisations to contact Genomics England was to question the exclusion of certain conditions which they felt met the selection criteria. Support organisations reported varying levels of involvement in the design of the Generation Study and the opportunity to comment on the 'Condition Suspected' information sheets which would be provided to parents. Some stated that they would have preferred earlier or more involvement in the Generation Study and welcomed future opportunities to engage with Genomics England.

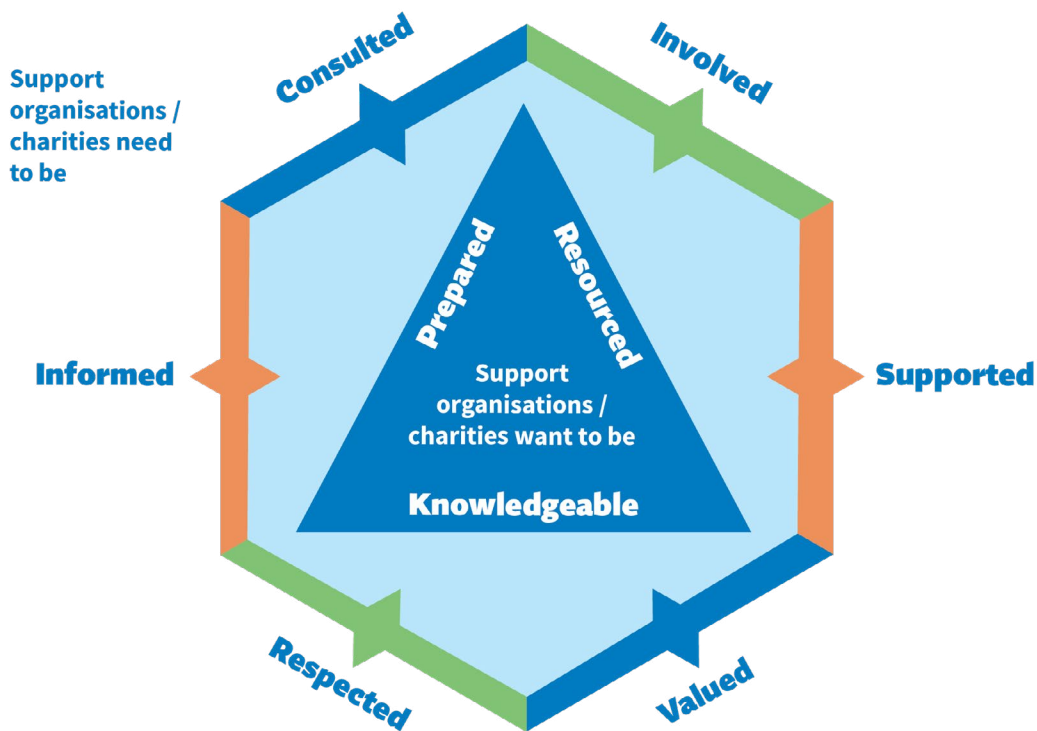
There were varying levels of knowledge about the Generation Study amongst the respondents with some wanting more knowledge around topics such as the selection criteria and how conditions could still be included within the Generation Study. Overall, respondents felt that there would not be much change in demand for their services because of the Generation Study. How prepared organisations felt to receive contacts from participants in the Generation Study depended to some extent on knowledge levels. It was felt by some that a lack of knowledge may reflect limited time and resources within their support organisation.

Support organisations generally wanted more information in areas where their knowledge was limited, this was particularly the case for those support organisations who were not familiar with having contacts from families where a newborn had a diagnosis, potentially without any symptoms. Respondents wanted resources and support in order to be able to give the best support to those parents / families who had participated in the Generation Study. Respondents felt that the responsibility for providing information and emotional support for those affected by a diagnosis sat with NHS England / Genomics England as well as support organisations and charities.

Support organisations identified many areas where participants in the Generation Study would need information and support. Respondents noted that it was important to follow up on the participants in the Generation Study and provide them with the necessary support for the long term. There were increased expectations for what the Generation Study can yield in the future and how it could become standard care with the potential expansion of the condition inclusion criteria. Respondents highlighted the continuing importance of careful management of people's genomic data and health information.

The experiences around the instigation of the Generation Study have highlighted what support organisations want to be (knowledgeable, prepared and resourced) and what they need from external players in order to be in this position (informed, supported, consulted and involved); also overall in the rare condition landscape the support organisations want to be valued and respected.

What support organisations need and what they want



Explanations of words / phrases used in this report

The Generation Study - A research study run by [Genomics England](#) in partnership with [NHS England](#). The study uses a single sample from a newborn to look for changes in genes linked to more than [200 rare conditions](#) to facilitate earlier identification. [The Generation Study](#) also aims to gather genomic data for wider research purposes. It aims to recruit 100,000 newborns in England.

The Generation Study Process and Impact Evaluation - [Genomics England has funded the Process and Impact Evaluation Research Team](#) to undertake an independent mixed-methods evaluation of the Generation Study. The team is made up of researchers from [UCL](#), [Breaking Down Barriers – Alström Syndrome UK](#), [Genetic Alliance UK](#), [Great Ormond Street Hospital for Children](#), [Queen Mary University of London](#) and [Warwick Medical School](#).

‘Included’ - Support organisations working with conditions which were included in the Generation Study list as of August 2024 – could be all or some of the conditions they support.

‘Excluded’ - Support organisations working only with conditions which were not included in the Generation Study list as of August 2024, or this is unknown.

Organisation - Charity or patient support organisation which works within the rare, genetic or undiagnosed sector, either directly supporting those with conditions and their carers or indirectly supporting by taking another role in the rare space.

Respondent - Respondents are the representatives from the charity / patient support organisations who took part in the [Generation Study Process and Impact Evaluation](#).

Participant - Participants are those who took part in the Generation Study.

Preparedness - How prepared support organisations think they are for being contacted by someone as a result of the Generation Study.

Condition suspected - One of the gene changes linked to one of the genetic conditions in the Generation Study was found from the blood sample. The parents are then informed of this result by telephone.

gNBS - Genomic newborn screening.

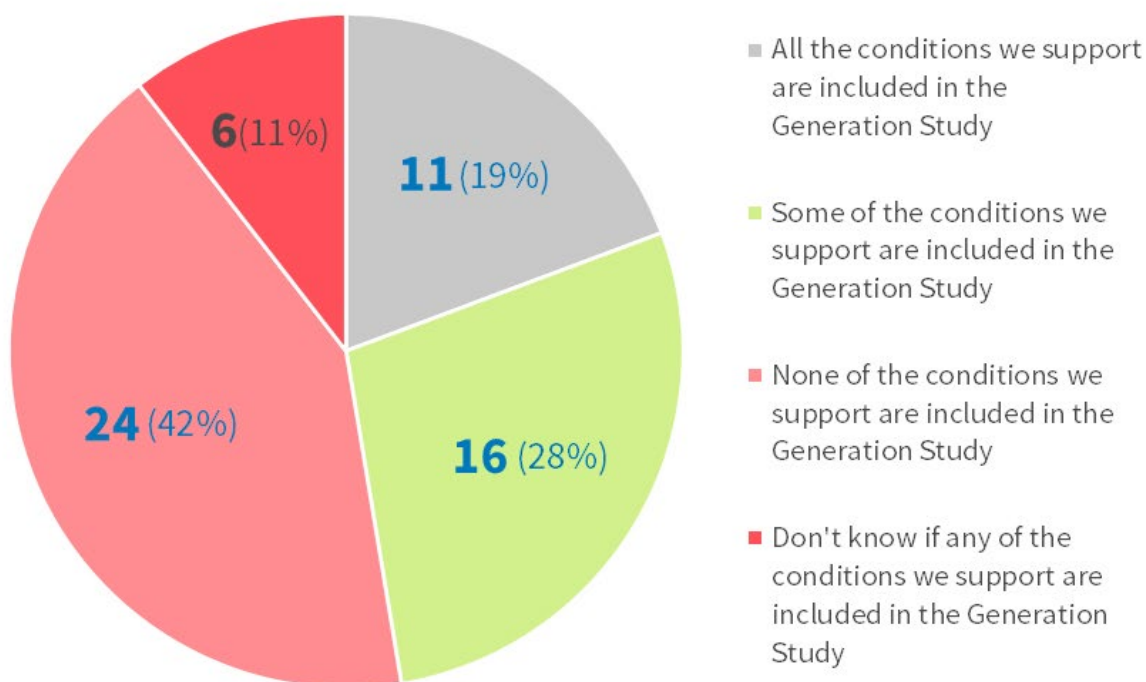
Introduction

The Generation Study is a research study run by Genomics England in partnership with NHS England to explore the benefits, challenges and practicalities of offering genomic newborn screening to parents in England. The study aims to recruit 100,000 newborns and use a single sample from the newborn to look for changes in genes linked to more than 200 rare conditions to facilitate earlier identification. Genomics England funded an independent evaluation of the Generation Study (the Generation Study Process and Impact Evaluation), which is made up of researchers from multiple organisations, including rare condition support organisations. The Evaluation considers a range of stakeholder views including parents, professionals, patient organisations and the public.

This report focusses on the attitudes of the rare condition support organisations and their views on the Generation Study. This work was predominantly undertaken by Genetic Alliance UK with support from the rest of the Process and Impact Evaluation team. We surveyed 59 support

organisations and charities in autumn 2024 and conducted 20 semi-structured interviews with advocates from those organisations between December 2024 and February 2025 (this was within the first six months of recruitment to the Generation Study). Of the 59 organisations, 57 directly supported people with rare conditions and were asked whether they supported any conditions included for screening in the Generation Study; results are shown in Figure 1.

We use 'included' in charts in this report to refer to support organisations working with conditions on the Generation Study list, as of August 2024 (27 respondents), and 'excluded' to refer to those only working with conditions not on the list or where this is unknown (30 respondents). A range of support organisations took part in the Evaluation. Around two thirds (67%) of the organisations who responded to the survey had two or fewer paid employees and 55% had an annual income below £100,000. Most organisations (91%) had volunteers working for them.



Those organisations who directly support (n=57)

Figure 1: Whether any of the conditions the organisation supports are on the list of conditions that are being screened for in the Generation Study

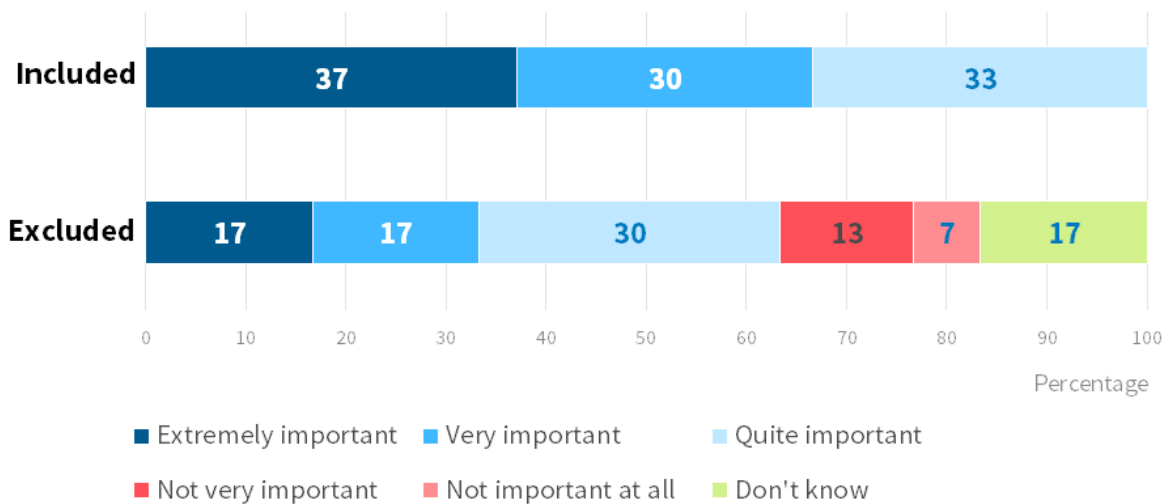
Evaluation findings

1. Communications from and engagement with Genomics England

Organisations taking part in the Evaluation study reported different levels of communication from and engagement with Genomics England. Some had been involved in working groups or had been included in consultations on the 'Condition Suspected' Information Sheets for parents whose baby had a suspected condition; while others had heard about the study but were not directly involved in any way. On the whole, respondents wanted to be informed about and involved in the Generation Study, and those with included conditions generally felt more strongly about this. Respondents also wanted Genomics England to be contactable and responsive to queries.

1.1 Being well informed about the Generation Study

Respondents wanted to be well informed about the Generation Study. In the survey, two-thirds (67%) of organisations supporting conditions in the Generation Study ('included') said it was extremely or very important to be well informed about the study; the remaining third (33%) considered it quite important (Figure 2). For organisations not supporting included conditions ('excluded'), a third (34%) rated being well informed as extremely or very important.



All respondents who directly support (n=57)

Figure 2: How important it is to the function of the organisation to be well informed about the Generation Study. Findings are shown by whether the organisation supports conditions included in the Generation Study or not.

Support organisations expressed a strong interest in regular updates on the Generation Study, including recruitment numbers and how many babies receive 'condition suspected' results. Information from Genomics England, through newsletters, webinars and other channels, is generally well received, though respondents suggested the website could provide more detail. They also wanted clearer explanations of each stage of the study, from enrolment to diagnosis,

to help them prepare and signpost families more effectively.

“ They have recently started a newsletter...I think it is specifically for patient organisations – which I think is brilliant and I think that’s very much needed and maybe ideally would have been started at an earlier timepoint than recently. I think people are very keen to hear how the study is going. ”

- Interview ID 2

Nearly all organisations that responded to the survey expressed the need to find out more about the Generation Study (93%). Their preferred routes are summarised in Figure 3.

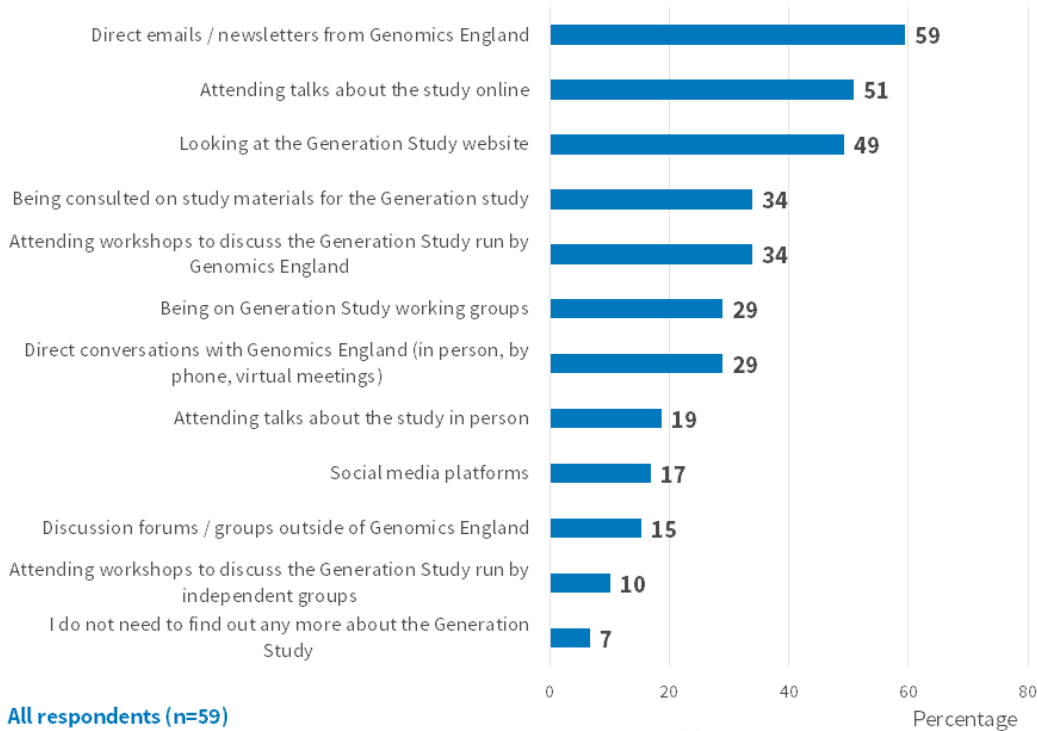


Figure 3: Ways in which organisations would prefer to find out more about the Generation Study

All respondents (n=59)

Support organisations want more information on:

- How conditions were selected for inclusion and how new conditions may be included
- Updates on the study's progress:
 - how many have been recruited, tested, found positive (especially for the relevant condition)
- More details on what happens at each stage of the study including:
 - recruitment, taking the sample, accuracy of the testing, technology used, further testing, treatment pathway, provision of support for the Generation Study participants
- Explanations of false positive and false negative results
- Better understanding around how genetics impacts treatment
- Details about the limitations of the Generation Study
- Wider implications of the Generation Study such as:
 - testing existing siblings, PGD for future pregnancies, the impact on data linkage to registries, demand to access data for future research, future of newborn screening
- The Generation Study outcomes and findings

1.2 Responsiveness of Genomics England to support organisations

Respondents reported mixed experiences with Genomics England’s responsiveness with some feeling heard and valuing positive communication and others feeling frustration over unanswered queries and limited early engagement. Some support organisations raised concerns about inaccuracies in the Generation Study ‘Condition Suspected’ Information Leaflets.

“ I’ve contacted the Generation Study team twice in May to have [name of condition] included in the list, but I have not received an answer ... I wish I would have received an answer to my email. I felt quite frustrated when I wasn’t able to receive an answer. ”
- Survey ID 15

“ We received emails from them as the patient advocacy groups involved ... our details were to be given to families. And we were given a chance to have a look at the information and the letters that would be sent out and the way it was phrased, and the support ... we were able to have an input into that. And that was good. ”
- Interview ID 8

“ When you ask for input...listen and act upon the input, or if not acting upon it be sure to feedback and explain why ... especially to those that took the time and effort to take part in GE consultations. ”
- Survey ID 20

Some support organisations questioned the exclusion of certain conditions they believed met the selection criteria. Several had raised

this directly with Genomics England, but said the rationale was unclear, unconvincing, or not provided. Some noted that these discussions were still ongoing. About two-thirds of respondents (66%) were unaware that Genomics England had a service desk available by phone or email for enquiries about the Generation Study.

“ It is easy to explain the selection criteria in respect of the four principles. It is impossible to explain why [name of condition] was not deemed to meet any of those conditions given that it could ‘tick the box’ for all of them comfortably. ”
- Survey ID 10

1.3 Levels of involvement with the Generation Study design

Levels of involvement varied with the design of the Generation Study. Just over half of ‘included’ organisations (56%) recalled receiving an email from Genomics England about the conditions they support being included, while 22% were unaware of any contact despite supporting included conditions (Table 1). Further, not all organisations were consulted on the development of the ‘Condition Suspected’ Information Sheet, and some said their feedback was either ignored or went unanswered.

“ I was concerned over the information leaflet that they sent me as draft. It was incorrect including on the passing of a [type of condition] from mother to child. I emailed by return and have had no (zero) response. I have also not seen the final leaflet. ”
- Survey ID 38

Table 1: Whether organisation is named as a source of support / information on any of the study documents. Findings are shown by whether the organisation supports conditions included in the Generation Study or not.

	Included	Excluded
Yes, we are named and have seen the study documents	41%	0%
Yes, we are named and but have not seen the study documents	11%	0%
No, we are not aware that we are named on any study documents	15%	37%
Don’t know	26%	30%
Other (specify)	4%	0%
Not applicable	4%	33%

Genomics England collaborated with support organisations in several ways. Nearly two in five respondents (39%) shared that their organisation had contributed to consultations on the parent ‘Condition Suspected’ Information Sheets. Input to other tasks was far lower, for example: working groups for designing / delivering the Generation Study– working groups included ‘Education and Training’, ‘Ethics’, ‘Evaluation’, ‘Recruitment’ and ‘Communicating Results’ - (19%), recruitment materials and the Generation Study website (7%), guidelines for delivering ‘condition suspected’ results (5%), and the ‘no condition suspected’ and ‘sample failure’ letters (3%). Many respondents (36–76%) were unaware of these opportunities.

Only 11% of organisations with included conditions felt they had been highly involved in the Generation Study’s design, and 3% among those with excluded conditions (Figure 4). Among organisations with included conditions, a third (33%) felt they had been involved as much as they wanted. For organisations with excluded conditions, only 10% felt sufficiently involved (Figure 5).

“ Probably as involved as I wanted to be, you know, there was opportunity, like I say, to ask questions, particularly during those workshops. And then again subsequently. ”
- Interview ID 19

Some support organisations said they would have preferred earlier involvement in the Generation Study’s design, particularly in decisions about condition selection. However, they also noted that their own limited resources and capacity were barriers, and acknowledged the difficulty of involving the large number of rare-condition organisations.

“ Some of the engagement sessions took place in person in East London starting in the morning. We are a small organisation and organising sessions this way is very expensive and burdensome for volunteers with caring obligations. ”
- Survey ID 29

“ I would have engaged with the study had I known about it at the early stages. ”
- Survey ID 44

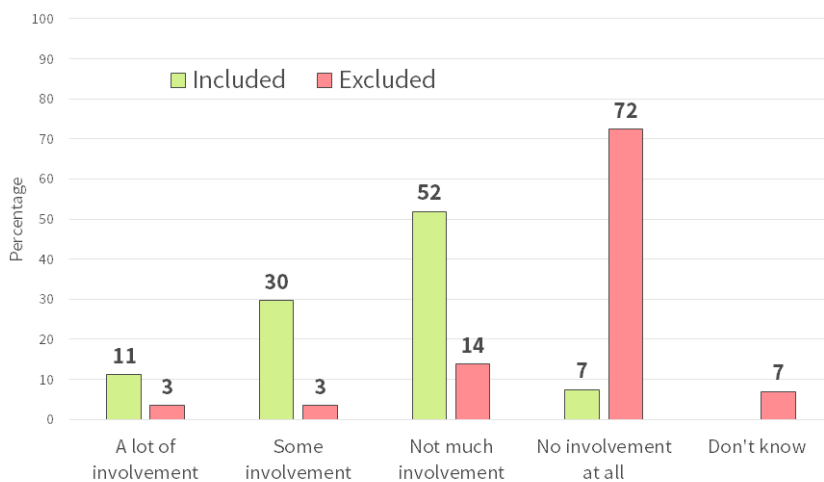


Figure 4: Level of involvement that the organisation felt it had in designing the Generation Study. Findings are shown by whether the organisation supports conditions included in the Generation Study or not.

All respondents that directly support and answered question (n=56)

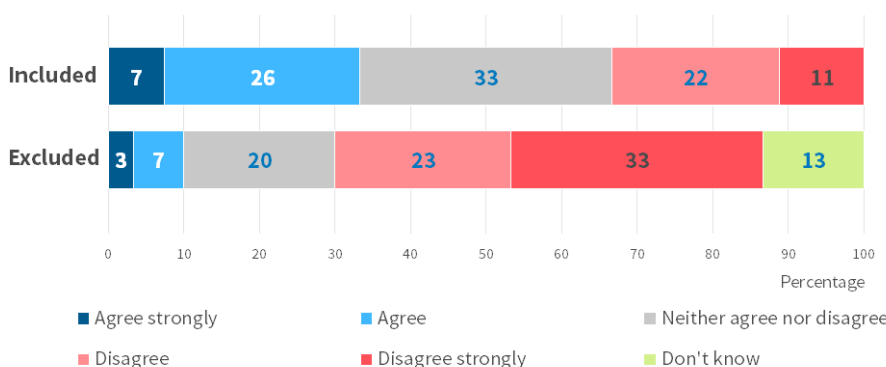


Figure 5: Agreement with whether the organisation had as much involvement in designing the Generation Study as it wanted. Findings are shown by whether the organisation supports conditions included in the Generation Study or not.

All respondents who directly support (n=57)

1.4 Onward engagement by support organisations

Engagement between support organisations and their communities about the Generation Study was limited, though some maintained broader conversations about newborn screening. Some respondents supporting conditions not included in the study said they had delayed discussions with their communities until talks with Genomics England were complete, to avoid raising false hopes.

“ No [have not discussed with community], I didn't want them to get their hopes up. I would rather just speak first with Genomics England to see if they were able to include this in. ”

- Interview ID 3

“ ... informed community about inclusion. Overwhelming positive response as many have been diagnosed later in life having gone through long diagnostic odysseys. ”

- Survey ID 43

Some respondents were concerned that included conditions might attract more research attention than those excluded, potentially widening existing inequities among rare conditions.

1.5 Future dialogue with Genomics England

Support organisations want their views represented across a range of issues and welcome future opportunities to engage with Genomics England. Issues include how Generation Study participants will be supported, with smaller, less-resourced organisations expressing concern that they have limited capacity to cope with additional workload if specialist centres do not meet support needs.

“ We would like to be involved in further conversations about embedding psychologically informed care for those taking part in this study, and how this could be better implemented in the NHS genomic services. ”

- Survey ID 49

Support organisations want to discuss these issues with Genomics England

- how conditions could still be included – how evidence on the decision-making process can be contributed to and how support organisations can call for condition decisions to be reconsidered
- how the mental health needs of the Generation Study participants are being addressed through the study design and the NHS
- what implications there are for potential increased workloads for some support organisations and how the Generation Study / Genomics England will support those organisations
- what experiences other support organisations have had if anyone has been in touch as a direct consequence of the Generation Study and how these learnings could be shared

Support organisations are keen to have their voice heard and get involved with Genomics England in future initiatives.

2. Knowledge levels and preparedness of organisations for the Generation Study

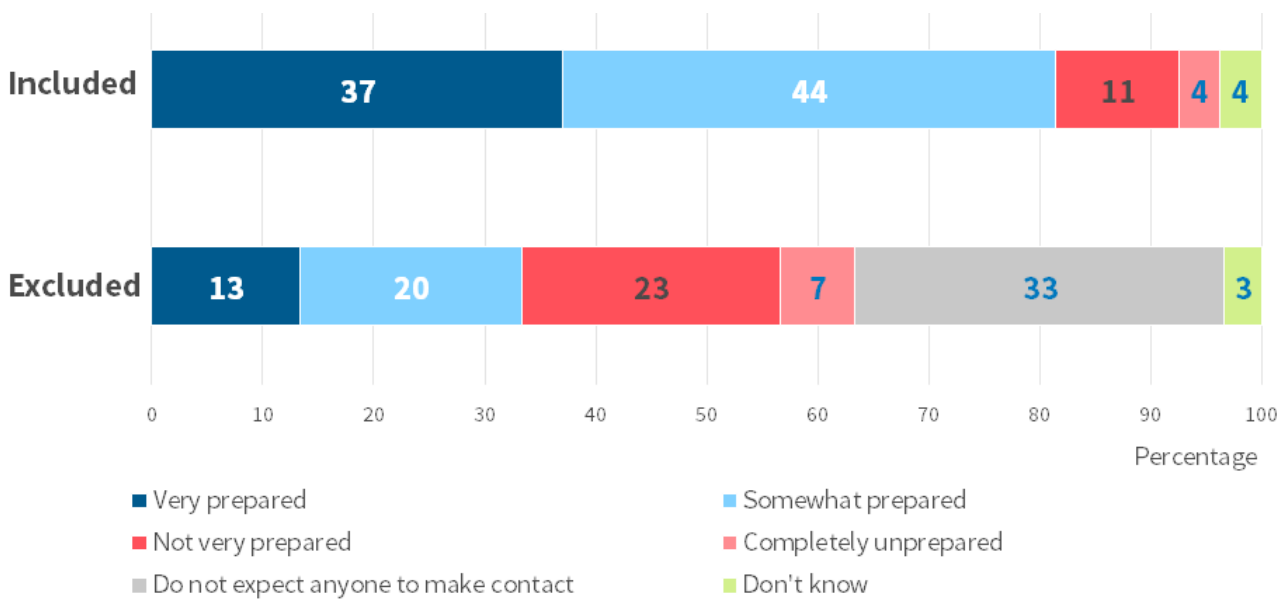
Support organisations described different levels of knowledge about the Generation Study and preparedness for supporting people who had participated in the Study. There were certain aspects of the Generation Study where respondents felt knowledgeable, such as understanding the selection criteria for the conditions. However, some support organisations still wanted more knowledge around the criteria used to select conditions and how conditions could still be included. Although overall they did not expect much change in the demand for their services, support organisations still wanted to feel prepared.

2.1 Level of current preparation

Preparedness generally reflected how much organisations knew about the Generation Study. Overall, just over half of surveyed organisations (54%) felt very or somewhat prepared to support someone contacting them about the Generation Study. But ‘included’ organisations are different to ‘excluded’ organisations: 81% of organisations supporting included conditions felt prepared, compared with 33% of those supporting conditions not included (Figure 6).

“ I have not had to explain it to anyone in simple terms as of yet, but I feel that as the study progresses and if/when we receive questions/calls about it, should this question be asked we would be able to engage in conversations/explanations about it. ”
 - Survey ID 13

Those with included conditions, or with some involvement in the study’s design and development, reported a broader understanding of its key elements and felt confident they were well informed.



All respondents who directly support (n=57)

Figure 6: How prepared organisation feels to support someone who contacts them after participating in the Generation Study. Findings are shown by whether the organisation supports conditions included in the Generation Study or not.

2.2 Knowledge about the Generation Study

The survey also showed variation in organisations' knowledge of the Generation Study's specific processes (Figure 7). Overall half of the respondents felt they had a lot or some knowledge of how conditions were selected (59%). Fewer than half of respondents felt that they had a lot or some knowledge around: how a blood sample was taken (42%), how parents will be recruited (39%), how blood samples will be tested (37%) and how 'condition suspected' results will be returned to parents (34%).

Finally, fewer than a third of respondents felt they had a lot or some knowledge about: the next steps for those who have a suspected condition result (31%), the long-term storage of genomic data (31%), and the use of genomic data for future research (29%).

Organisations reported the need for more knowledge in the areas they felt least knowledgeable. The notable exception was for 'How conditions were selected' – overall 41% still said they would like more information about this. However, within this number there is a wide

discrepancy between those who had conditions included where 22% wanted more information, and those who did not support conditions included where 57% wanted more information (Figure 7).

“ Hopefully I do feel pretty well informed about it. It is an evolving picture as well and my understanding is always developing. And that continues. But I suppose I am in that position of I've had opportunities to talk at times with people from Genomics England who are organising the study and to attend certain meetings and things, which has given me information about how the study's working and how it's going and things. ”

-Interview ID 2

“ I wouldn't say I was that knowledgeable because I have so much work to do. ”

- Interview ID 18

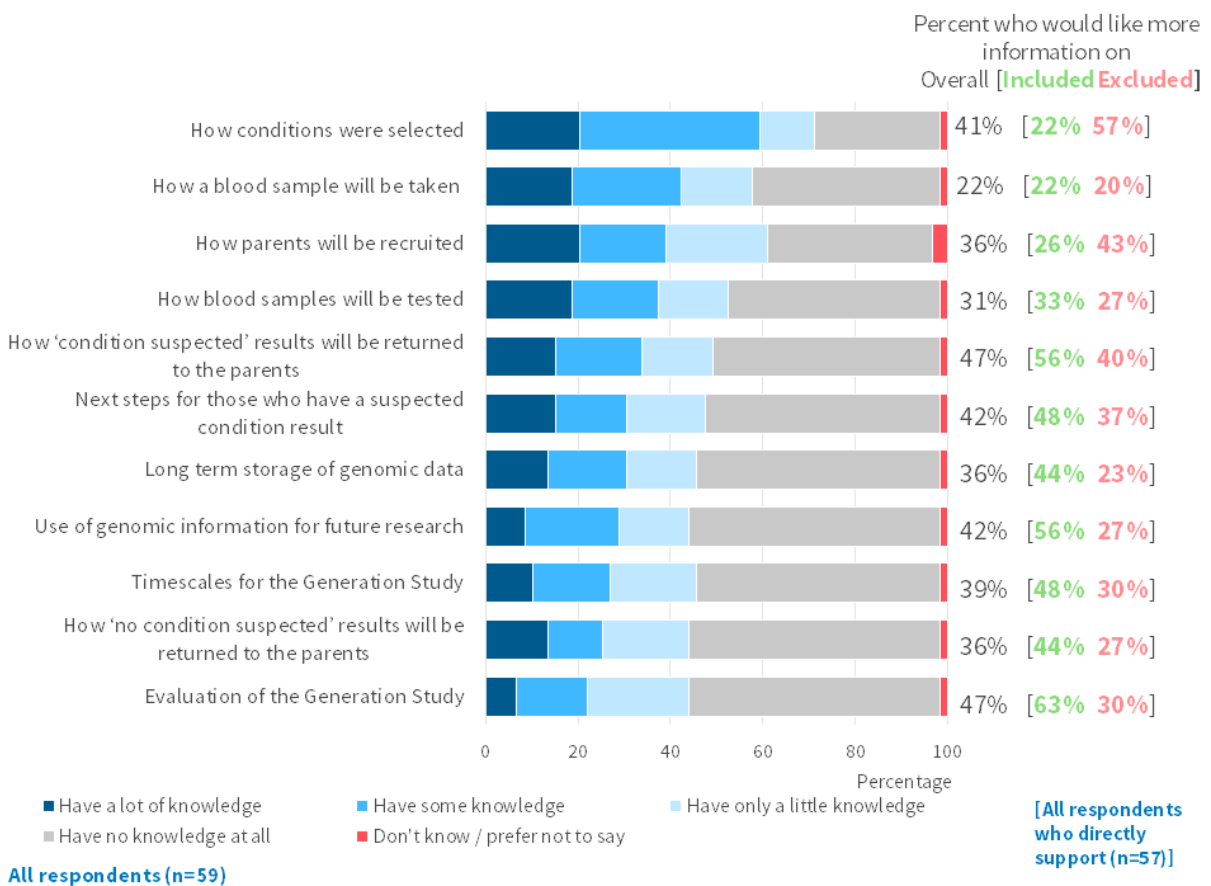


Figure 7: Knowledge levels organisation feels they have about the different aspects of the Generation Study and what they would like more information on. Findings are shown by whether the organisation supports conditions included in the Generation Study or not.

2.3 Knowledge about the selection criteria

Just over half of respondents (53%) felt confident they could explain to their community how screening conditions were chosen, while more than a quarter (28%) were not confident.

“ So I understand that effective treatments is part of it. And I think I remember reading about that the accuracy of diagnosing through the test would need to be robust, so conditions that maybe could or couldn't be might not be chosen. There's others as well but I can't remember what they are.

- Interview ID 20

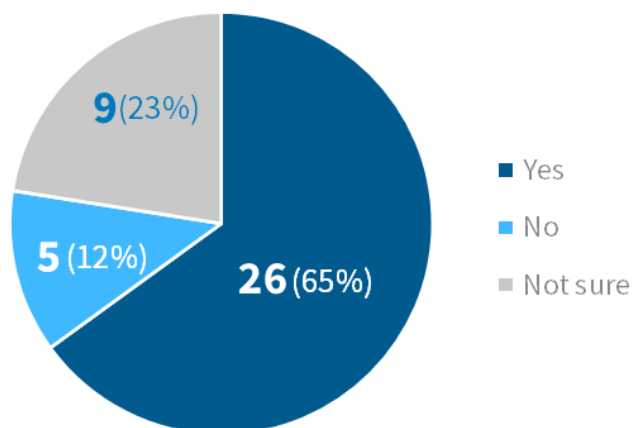
Some respondents were unclear about how the treatable / early intervention inclusion criterion had been applied, noting that many conditions have symptoms that can be managed to improve quality of life. They also emphasised that early diagnosis offers benefits such as better understanding of the condition and access to peer support.

“ I think this idea about treatability is quite tricky ... because often we know there is no treatment for lots of conditions, but there are things you can do, you know, for the developmental conditions getting early intervention we know is really important and effective.

- Interview ID 5

2.4 Future of further inclusion / deselection possible

Among organisations with none or only some of their conditions included (40 respondents), around two-thirds (65%) said they would have liked more conditions included, and about a quarter (23%) were unsure (Figure 8).



Those organisations who said 'some' or 'none' of the conditions directly support are on the conditions list (n=40)

Knowledge varied regarding the possibility of adding or removing conditions during the **Generation Study**. Organisations without included conditions welcomed the potential for future inclusion but were unsure whether they could nominate conditions, and there was uncertainty about why conditions might be deselected once the study had begun.

“ It's good that new ones can be included. I'm not sure how I feel about stuff being removed. I think there needs to be a little bit more detail around that. It would seem pretty harsh if you were in there being screened and you got removed. There would have to be very good reasons for it.

- Interview ID 14

“ Our understanding is that more conditions might be added but are unclear about the timeline for this and how the process for doing so works, e.g. can support groups put forward a condition for consideration?

- Survey ID 24

2.5 Changes in demand for services

Overall support organisations did not expect there to be much change in demand for their services due to the **Generation Study**. Table 2 on the following page shows expected changes in demand for organisations' services following diagnoses from the **Generation Study**. Only 4% of 'included' organisations anticipated a lot of extra demand and 41% expected a little extra demand. Among 'excluded' organisations, 20% expected a little extra demand.

Figure 8: Would have liked the condition(s) supported (or more of them) to be included in the **Generation Study**

Table 2: Expected changes in demand for the organisation’s services as a result of newborns being diagnosed through the Generation Study. Findings are shown by whether the organisation supports conditions included in the Generation Study or not.

	Included	Excluded
Yes, I expect there to be a lot of extra demand because of diagnoses made by the Generation Study	4%	0%
Yes, I expect there to be a little extra demand because of diagnoses made by the Generation Study	41%	20%
No, the organisation is not expecting any changes in demand	22%	60%
Difficult to say / it is not known whether there will be any extra demand for the organisation	33%	20%

Very few respondents were aware of any direct contact resulting from a Generation Study diagnosis or related queries. One organisation reported a small number of enquiries from people asking why certain conditions were not included.

“ I think we are geared up OK... so I may be wrong, but I imagine the increase in demand will start off relatively slowly and then gather a considerable amount of momentum, so we’ll have that rolling stone effect which will give us time to gear up. ”
- Interview ID 15

3. Information and support needs

Organisations were asked about the information they would like to have about the Generation Study and what type of support would be most helpful to them to be able to support participants. Support organisations also outlined what support and information they thought families who participated in the Generation Study would need if a newborn received a diagnosis.

3.1 Organisations’ views on what information they need

Some organisations unfamiliar with supporting parents of newborns with a diagnosis said they would need training or expert guidance on how best to support these families. Organisations said they would seek support from clinical experts and other experienced organisations to develop new resources for parents, but acknowledged limits in their own capacity and resources.

“ So we’ve been quite well included from day one. I think we still have perhaps a few unanswered questions, feel a bit under-equipped with supporting these families of, parents of newborns with apparently healthy newborns, to whom it will be a shock to have the diagnosis for those who don’t already have a diagnosis within the family. ”
- Interview ID 11

“ I am thoughtful as to how much has been taken into account about the psychological impact over time. I don’t know what funding has been allocated or is being looked to, for the future, to conduct those studies, and more importantly not just study, but put support in place for the longer term... So I think like many small organisations, the question we have will be around funding for capacity. We think we’ve got a lot to offer, but of course it will hinge on that. ”
- Interview ID 16

“ When it’s that first shock diagnosis with a newborn, parents have no idea what to expect. Will they be walking, talking like healthy toddlers? They just wouldn’t be able to picture it I don’t think without having prior experience of it. I think that actually the most helpful thing to us has been other charities who have reached out to say “We have experience of supporting parents of newborns who have been diagnosed through the heel prick screening” ... and they’ve told us how they have packs on their website specifically for parents in that situation, guidelines for the charity of how to support these families. So really that’s been the best help we’ve had so far.

- Interview ID 12

Support organisations / charities would like this type of support / information from Genomics England

- Genomics England to provide the full documentation pack which would be given to the parents with a suspected diagnosis so support organisations are familiar with the contents
- Email / telephone prompt guide designed with Genomics England to be used by support organisations if contacted by someone who had taken part in the Generation Study
- Training / expert advice on speaking to parents of newborns receiving a diagnosis for support organisations who were not familiar with providing this support for early diagnosis
- Support with resources and direct funding such as help producing and printing an information leaflet for parents who received a condition suspected letter
- Provision of easily accessible resources about the Generation Study in the form of leaflets, factsheets, videos, FAQs, a breakdown of some of the medical jargon, information about the genetic types and how this guides treatment – could be located on the Generation Study website
- Regular updates about the Generation Study in lay language / social media posts which could be disseminated to the support organisations’ members and community

3.2 Support for organisations

Some organisations said they would like more support from Genomics England to prepare for parent enquiries, including training and guidance. Access to the full information pack given to parents, along with factsheets and videos that could be shared on their own websites, was seen as helpful. Several also wanted links from the Generation Study website to their own website and suggested the ‘Condition Suspected’ Information Sheets could be hosted on the Generation Study website. They would welcome Genomics England staff attending their webinars, podcasts and conferences.

Many organisations noted they would need additional resources if the Generation Study increases their workload, including funding, staff time, external costs and meeting spaces.

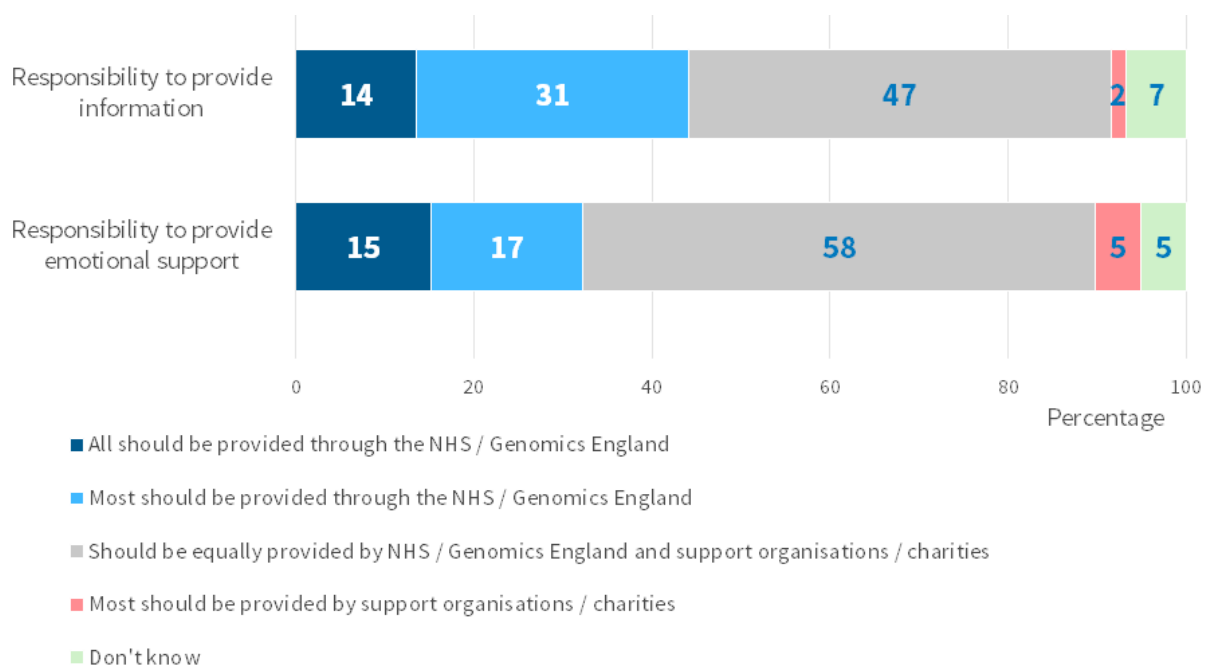
“ A complete pack of information provided to the parents so we are aware what information they are given so we can respond appropriately. ”
- Survey ID 55

“ Resources would be great. Maybe guidance on the best way to approach these topics, especially if we’re engaging with parents who are concerned about ... how best to handle sensitive topics ... And yeah, I think resources would be great, things that we could share on our website. ”
- Interview ID 9

3.3 Support for parents

Respondents wanted parents of babies diagnosed through the Generation Study to be signposted to relevant support organisations but most felt the primary responsibility for supporting families lies with the NHS and Genomics England (Figure 9). Demand for support group services was expected to grow over time while there were also concerns about whether the NHS and Genomics England could provide the emotional support families may need.

“ Unfortunately there is a complete lack of emotional support within the NHS, needs to improve. ”
- Survey ID 35



All respondents (n=59)

Figure 9: Responsibility to provide information / emotional support to those receiving a diagnosis through the Generation Study

Figure 10 summarises the information and support respondents felt parents would need when a baby receives a diagnosis through the Generation Study, including:

- Treatment and management options (86%)
- Connections with knowledgeable healthcare professionals or specialists (85%),
- What a suspected diagnosis means (80%)
- Connections with others for peer-to-peer support (78%)
- Potential outcomes for someone with a diagnosis (75%).

“ We would hope that NHS England / Genomics England are actively encouraging and sign posting affected families to contact reputable support groups. ”

- Survey ID 57

“ The responsibility lies with the state / official organisations, but the charities have a lot to offer. Some are better resourced than others, however. ”

- Survey ID 45

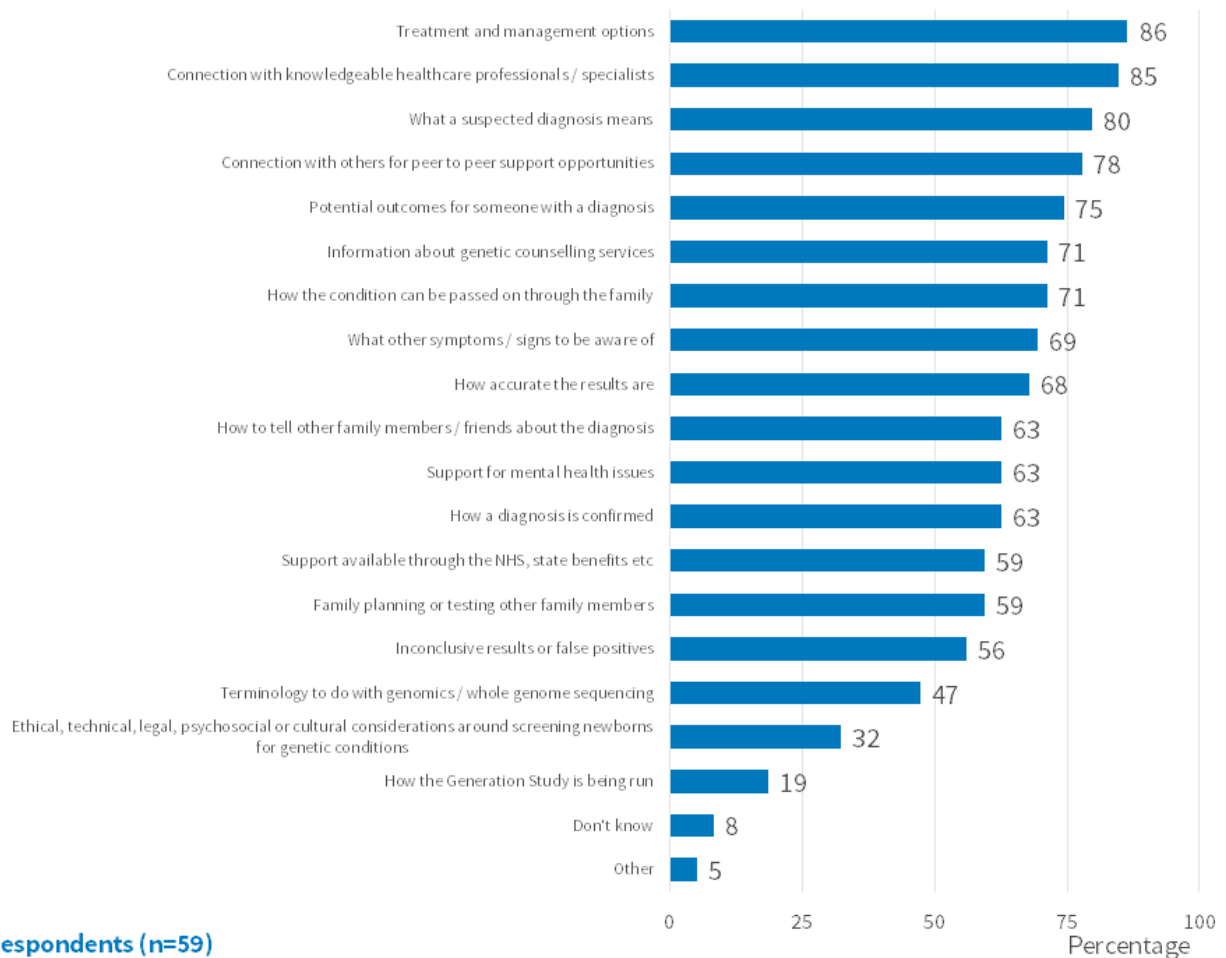


Figure 10: When a baby is diagnosed through the Generation Study, what organisations think will be the main support and information which parents / carers need

Recommendations

Use a variety of formats for communication with support organisations:

- direct emails and newsletters
- online talks
- Generation Study website kept up to date, with study information provided as leaflets, videos, and factsheets
- be responsive to specific queries made about the Generation Study

Ensure support organisations have access to:

- information about study design including how conditions were selected and how further conditions could be included
- information about the process for parents across the various stages of the Generation Study from recruitment through to returning findings and follow-up care
- the documentation which is given to parents when a newborn is diagnosed so support organisations are better prepared
- detail about how the genomic data collected will be stored
- information about how participants in the study will be provided with emotional support
- progress updates and final outcomes of the Generation Study

Initiate further discussions with support organisations on:

- how decisions around removal of conditions are made
- whether and how the organisations can request further conditions be considered for inclusion and how they can provide evidence into the decision-making process
- the future of gNBS shifting into routine care and other new initiatives from Genomics England / NHS England

Offer the following support to organisations:

- guidance and training around supporting families of newborns who receive a diagnosis
- resources and funding to help support organisations prepare and meet the needs of parents

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- Alport UK
- Alström Syndrome UK
- AMEND
- Angelman UK
- Annabelle's Challenge
- Beckwith Wiedemann Support Group
- Behçet's UK
- Brittle Bone Society (BBS)
- Cadasil Support UK
- Child Growth Foundation
- CSNK2A1 Foundation
- Cystinosis Foundation UK
- DBAS
- DEBRA
- Dystonia UK
- Ectodermal Dysplasia Society
- EURORDIS Rare Diseases Europe
- Fanconi Hope
- FOP Friends
- Gene People
- Glut1 Deficiency UK
- Headlines Craniofacial support
- Immunodeficiency UK
- ITP Support Association
- KBG Foundation
- Marfan Trust
- Matthew's Friends
- Max Appeal
- Medics for Rare Disease
- myaware
- National Society for Phenylketonuria
- NCBRS Worldwide Foundation
- Nephrotic Syndrome Trust
- Neuro Therapy Network
- Noonan Syndrome Association UK
- Pernicious Anaemia Society
- Phelan-McDermid Syndrome Foundation UK
- Pompe Support Network
- Rareminds
- SWAN UK (run by Genetic Alliance UK)
- The Children's Hyperinsulinism Charity UK and Ireland
- The Duchenne Family Support Group
- The Fragile X Society
- The UK Mastocytosis Support Group
- The Wilson's Disease Support Group - UK
- The XLP Research Trust
- Timothy Syndrome Alliance (TSA)
- TTP Network
- Turner Syndrome Support Society UK
- UKPIPS
- Unique
- Williams Syndrome Foundation

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