Spectrum 10K Consultation

Phase 3: the findings

What was said about how to improve Spectrum 10K

An independent report
Hopkins Van Mil and Karen Leneh Buckle

September 2023

Contents

Executive Summary

1. **Background:** This is the report of Phase 3 of the consultation on the Spectrum 10K autism genetics research study (‘the study’). The consultation was initiated in response to vocal opposition to the study’s launch in August 2021. It has been co-designed with autistic people. Phase 3 invited people from the autism community[1] to give their views on how the study could be improved to make it feel safer and more acceptable. Hundreds responded, most (70%) of them autistic. It uses qualitative methodology supplemented by some closed quantitative questions which are easier for some autistic people to answer.
2. **Spectrum 10K’s relationship with the autism community:** Opposition to the study was largely fuelled by the genetic nature of the research and by feelings about the research team. Consultees’ views range from admiration for the team and complete support for the study to strongly negative opinions about the team and feeling the study is beyond remedy. Many consultees welcome the consultation, although a few think it will not be sufficient to make real change to the study and others felt the study should not have been paused. The autism community is highly diverse and there is no united view on genetic research or Spectrum 10K even within subgroups of the community (e.g. autistic people who can speak for themselves).
3. **Aim to look for genetic contributions to autism:** Some consultees feel there has been a lack of clarity, openness and consistency about the aim to look for genetic contributions to autism and co-occurring conditions. Concerns about eugenics and prenatal testing that were raised after the study’s launch were a strong theme in the consultation responses. Some consultees were concerned the data could be used by other scientists in the future for eugenics or the development of a prenatal test for autism, and consultees feel there has been a lack of clarity about how this will be prevented. Other consultees welcome research into the genetic basis of autism and co-occurring health conditions.
4. **Further aims and potential benefits:** The study has further aims to look at genetic underpinnings of health and wellbeing. Some consultees feel the path from basic research to future benefits is unclear. Other consultees support the aim to look at the genetic basis of co-occurring health conditions. Some consultees are particularly concerned about the aim to look for genetic subgroups because they believe that subgroups have been harmful to autistic people in the past. While most consultees support research into co-occurring conditions, there are questions about why a genetic approach is necessary.
5. **Data collection and security:** Data collection using questionnaires is relatively uncontentious amongst consultees, if adapted for autistic processing. Access to medical records was initially a compulsory aspect of the study, but the proposal to make this optional is widely supported by

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[1] Throughout this report, ‘autism community’ is used to refer to autistic people, family members, and others with a personal connection with autism, whether they are members of any autism-related groups or communities or or not.

consultees. Some consultees are concerned about medical records not being a true representation of the person. There are further concerns that the data could be leaked or may be improperly anonymised. Many consultees are satisfied that the planned robust data security measures will be sufficient protection. A few consultees support compulsory inclusion of the sharing of medical records.

6. **Consent and withdrawal:** Some consultees are supportive of including children and adults without capacity to consent to enable a diverse dataset, while others question the ability of non-autistic people (e.g. parents) to determine the interests of autistic people. Some consultees ask for robust assent procedures and safeguards to confirm consent at different points in the study. They also request fine-grained consent for components of the study and data sharing. The proposed simplification to clarify the withdrawal procedure is largely supported by consultees.
7. **Ongoing engagement:** Consultees say that autistic people must be involved throughout the lifespan of the study. Ongoing open communication is needed. They ask researchers to enable access by as many autistic people as possible by providing multiple formats of communication and engagement.
8. **How to improve Spectrum 10K:** The purpose of the consultation was to engage with the autism community to find out what they would like to change about the Spectrum 10K study in order to improve it. Researchers are encouraged to consider all of the recommendations and all the voices heard during the consultation. Some consultees wonder whether, in the face of strong objections, genetic research is in the interests of the autism community at this time. The consultation demonstrates the great diversity in the autism community about this research study. Long term engagement with community interests, priorities and objections is encouraged.

Summary of recommendations

A full list of recommendations can be found in Appendix 1. A summary of the key recommendations is provided here.

Provide clear, consistent and detailed information

Aim to balance detail and clarity in information without compromising integrity. Autistic people are highly sensitive to inconsistencies. Respond openly and honestly to questions and challenges as they arise. Honest answers should not take inordinate time to produce**.** Continue to communicate about ongoing developments, including the outcomes of the consultation, discussions with the Health Research Authority (HRA), and future uses of Spectrum 10K data.

Study documentation should be produced in multiple formats e.g., Easy Read to facilitate access. Be publicly positive about autism and use positive framing in documentation. Avoid words like ‘risk’ or ‘causes’ in the context of autism and be clear about what is meant by ‘treatment’.

Involve autistic people with a variety of views and experiences

Continue to consult with, include, and involve autistic people in the governance of the study, as members of the research team and advisors, and to review study materials and documentation. Seek to include the widest possible range of views and experiences, including those who are critical of genetic research and those who are negative about being autistic. Apologise for insensitive and/or disrespectful comments. Consider the research priorities of the community and spell out clearly the path from basic research to future benefit.

Allow individual control wherever possible

Autonomy provides opportunities for confidence and trust. Wherever possible, facilitate control over elements of the study participants wish to engage with. As much as possible should be optional. Explaining the reasons for requested information will encourage participants to make informed choices.

Take strong steps to prevent misuse of data

Acknowledge the fear that genetic research may lead to eugenics applications. Be honest about the risks of unintended uses of published data, and take concrete steps to reduce them. Convene a **Data Access Committee (DAC)** giving a controlling majority and/or veto to autistic members. Develop guidelines for data access that include adherence to ethical guidelines, proper use of data and the interests of autistic people.

Use robust and accessible methods for consent and withdrawal

Ensure that descriptions of possible benefits and risks of the research are clear, balanced and provided before consent is obtained. Simplify the withdrawal process, but retain an option to withdraw only from contact. Provide information and recording of assent in different formats for accessibility, and require assent from all children with the ability to understand the adapted study information.

Part one: Background to the consultation

This report is in eight sections, which are divided into four parts.

1. **Background to the consultation:** Why the consultation was launched, how it was done, and what we did in Phase 3 specifically. This part also includes some of the background that led to the study being paused to conduct this extensive consultation.
2. **Aims and intentions of the Spectrum 10K study:** Includes responses about what Spectrum 10K is or isn't aiming to achieve, including public concerns about prenatal testing.
3. **Study methods:** Data collection, storage, consent and withdrawal.
4. **Going forward:** Ongoing involvement of the autism community in Spectrum 10K, communication, and improving the study.[2]

As described in detail later in the report, the purpose of the consultation is to improve the study, and most of the report focuses on concerns from the autism community and suggestions for addressing those concerns. Nevertheless, it is necessary to acknowledge the responses from those who think the study should be stopped. These are presented at the beginning of the report in section two, before going into detail about how the study could be improved in sections 3-8.

1. The consultation

1.1. Background

Spectrum 10K is a research project led by the Autism Research Centre[3] at the University of Cambridge (referred to as 'the team' in this report). The study aims to investigate genetic and environmental factors that contribute to autism, co-occurring conditions, and the wellbeing of autistic people. In particular, the study aims to identify common genetic variants associated with autism, autistic traits, and chronic health conditions that occur more often in autistic people (such as epilepsy and gastrointestinal pain).

Within a week of the project launch in August 2021, thousands of autistic people or their parents signed up; however, simultaneously, vocal critics raised concerns about the study protocol and research team (see Chapter 2 for more detail). Although no breaches of the ethically approved study protocol occurred, the project team decided to pause the study, with the support of the Wellcome Trust[4] (the funders) and the University of Cambridge, to allow time for larger scale and

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[2] Throughout this report, 'autism community' is used to refer to autistic people, family members, and others with a personal connection with autism, whether they are members of any autism-related groups or communities or or not.

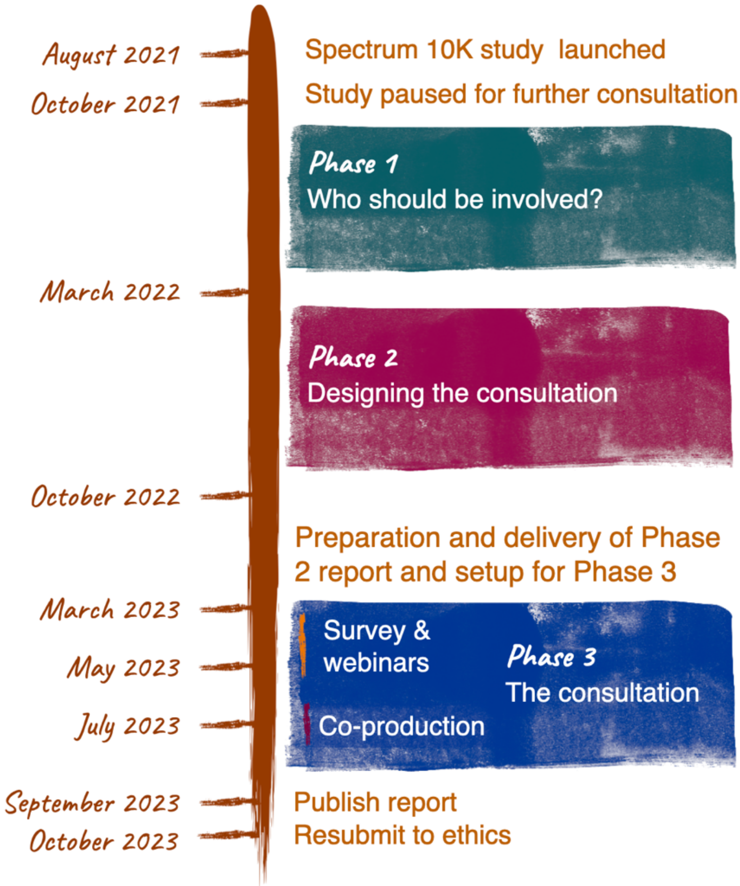
[3] https://www.autismresearchcentre.com/

[4] https://wellcome.org/

more diverse consultation with the autism community. The Wellcome Trust welcomed the consultation and provided additional funding to support it.

(See Figure 1 for timeline of events.) Although the team had consulted with a small advisory group comprising autistic people, parents of autistic children, and clinicians, the team recognised that with a project of this scale a larger consultation would have been better. The team decided to conduct a consultation with hundreds of people from the autism community, to better gauge their diverse views.

Figure 1: Timeline of Spectrum 10K from launch through consultation



The purpose of the consultation has been to enable the Spectrum 10K researchers to listen to opinions, ranging from fully supportive to deeply critical, and discuss the complex bioethical issues surrounding genetic research in autism, allowing Spectrum 10K an opportunity to learn from different perspectives, adapt the project, and build trust in the study. The hope is that the consultation will contribute to rebuilding trust between scientists and those who they believe might have a hidden agenda of eugenics, and to improve the study, and to set an example for the wider field of autism genetic research on how to conduct such research safely.

1.2. Who is running the consultation?

The consultation is led jointly by:

- Hopkins Van Mil
- Leneh Buckle
- Spectrum 10K research team

Hopkins Van Mil

Hopkins Van Mil (HVM) is a small social research and public engagement agency. It specialises in consultations which take time to engage with all those who need to be involved. HVM facilitates engagement so that voices are heard, learning is shared and understanding achieved. In October 2021 HVM was commissioned in a procurement process run by the Autism Research Centre at the University of Cambridge to work on the Spectrum 10K consultation. HVM's role in the consultation is to work as independent and impartial facilitators. This means we listen to and explore the perspectives of the autism community to find constructive ways forward.

Leneh Buckle

Leneh is an autistic researcher[5] and social care trainer with long experience working productively with neurodiverse teams through organising Autscape[6] and other projects. Leneh joined as a co-lead of the consultation alongside HVM and Spectrum 10K following recommendations from Phase 1 that an autistic person should lead the consultation. Leneh has been working on a freelance contract with HVM to facilitate the consultation co-design process and to interpret and report on the outcome.

Spectrum 10K team

The consultation is overseen by the team at Cambridge to make sure that the outcome is useful, relevant, and will help to address concerns and improve the study, and so that the team could participate in webinars to listen to and answer questions posed by consultees. The Spectrum 10K team will use the findings from the consultation to inform and adapt the Spectrum 10K study.

1.3. What is the consultation process?

The consultation had three phases. The timetable for the consultation is set out in Figure 2.

A report has been produced for each phase of the consultation and these are available on the Spectrum 10K and Hopkins Van Mil websites. These reports give recommendations which inform how the next phase of the consultation was conducted. We summarise key points below. The current report explains what people said in the consultation itself, which was called Phase 3.

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[5] http://autisticinertia.com/

[6] http://www.autscape.org/

Figure 2: Consultation timeline

Phase 1

Deciding who should be involved in co-designing the consultation

35 people (67% autistic) involved in small group discussions/ interviews

Completed March 2022

Phase 2

Co-designing the consultation

111 people (85% autistic) involved in small group discussions/ interviews/ work in a dedicated online space

Completed October 2022

Phase 3

The consultation

531 people (77% autistic) who responded to the consultation survey & took part in topic-based discussions.

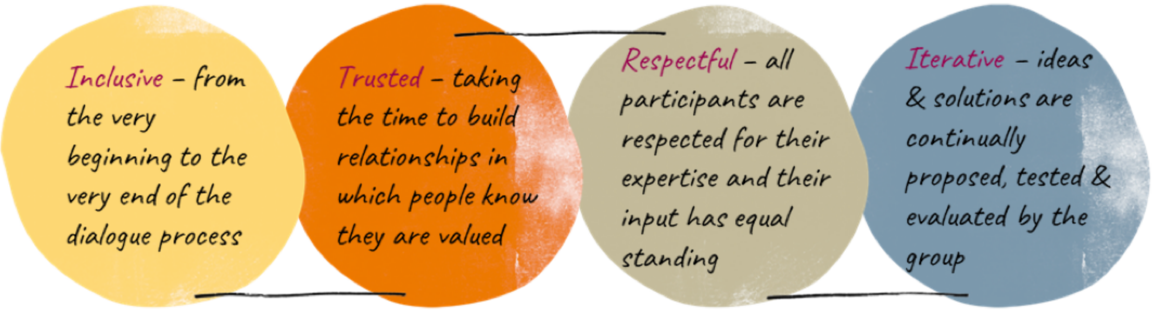
Completed May 2023

1.4. Phase 1 recommendations

The report from Phase 1 which focused on who should co-design the consultation was published in May 2022. An important recommendation from the Phase 1 discussions was the need to have an autistic person to lead the consultation. Because HVM did not already have someone on the team who could provide that, we ran a recruitment process to engage an autistic co-lead. This freelance appointment was made in July 2022.

We also learned from our discussions in Phase 1 that people highly value a process which is co-designed with autistic people throughout. Four principles discussed in Phase 1 therefore underpinned our work on this consultation. These are described in Figure 3.

Figure 3: The principles of co-design highlighted by Phase 1 co-designers



Inclusive – from the very beginning to the very end of the dialogue process

Trusted – taking the time to build relationships in which people know they are valued

Respectful – all participants are respected for their expertise and their input has equal standing

Iterative – ideas & solutions are continually proposed, tested & evaluated by the group

1.5. Phase 2 recommendations

Using the principles co-created in Phase 1, Phase 2 focused on co-designing the consultation. Co-designers advised the team on how Phase 3 of the consultation should run. Phase 2 also generated overarching principles for the consultation which were:

1. Build trust in the process
2. Make real and positive change to the Spectrum 10K study

3. Show that the consultation involves, and has been designed by, autistic people
4. Create a space for constructive discussion where people can hear, learn from and respond to a range of views.

There were specific recommendations for how the consultation should be designed which are set out in full in the Phase 2 report. In summary they are:

Timing

Co-designers recommended that the consultation should allow consultees enough time to reflect and process information, but not take so long that the process is too intense or for it to create anxiety.

Outcome: Phase 3 of the Consultation ran for 2.5 months from early March to the end of May 2023. There are limitations to how long the consultation could take due to the effect on funding for the study.

Information

The co-designers of the consultation said that people taking part in the consultation would need information provided by Spectrum 10K and by others with an interest in Spectrum 10K, including from those who have been critical of it. They said a clear statement of the study aims, objectives and planned outcomes was required, as well as other contextual information.

Outcome: The Spectrum 10K team provided a 36-page 'Spectrum 10K Themes and Responses to Questions' document. A summary of each section of this document was added to the HVM website. In addition, the consultation co-leads reviewed material produced by critics of Spectrum 10K and added summaries of these documents to the materials available to Spectrum 10K consultees. The main questions asked by critics and others unclear about certain aspects of the study were also added to the consultation page on the HVM website. This material also provided the preamble to each of the consultation questions.

Who should be involved in the consultation

Phase 2 co-designers recommended an inclusive approach to the consultation to enable all those who wished to share their opinions on how to improve and change the Spectrum 10K study to take part.

Outcome: The consultation was available in multiple formats. There were standard and Easy Read[7] questionnaires in the Qualtrics survey platform. Five webinars during which members of the public could ask questions of the research team were also held as part of the consultation. In order to gather responses from people who may find answering open-ended questions difficult, the surveys contained both closed questions (e.g. "How much do you agree with…?") and space for free text responses.

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[7] Thank you to A2i for the Easy Read version www.a2i.co.uk (reference 36543)

To ensure that a range of views and backgrounds were included, consultees were asked about their connection to autism and their attitude towards genetic research and Spectrum 10K.

1.6. Phase 3: Aims

The consultation process

The need for an iterative, staggered approach to the consultation was expressed by Phase 2 co-designers. They said that it is important to give time for people to consider their responses, to deal with controversial issues, and build trust in the consultation.

Outcome: The survey responses were processed over time with webinars and drop-in sessions near the end.

Make real changes to the study

It was important to co-designers that the consultation not be tokenistic and make real change to the study.

Outcome: The final stage brings together a small working group (up to 10 co-producers) of autistic people who are paid for their time to work together with the Spectrum 10K team to create the detailed change and improvements to the study set out in Phase 3. This process includes:

- Up to four facilitated co-production workshops
- An online space for reviewing documents and discussing detailed points
- A smaller subgroup to review the Spectrum 10K website and registration process

1.7. Phase 3: Methodology

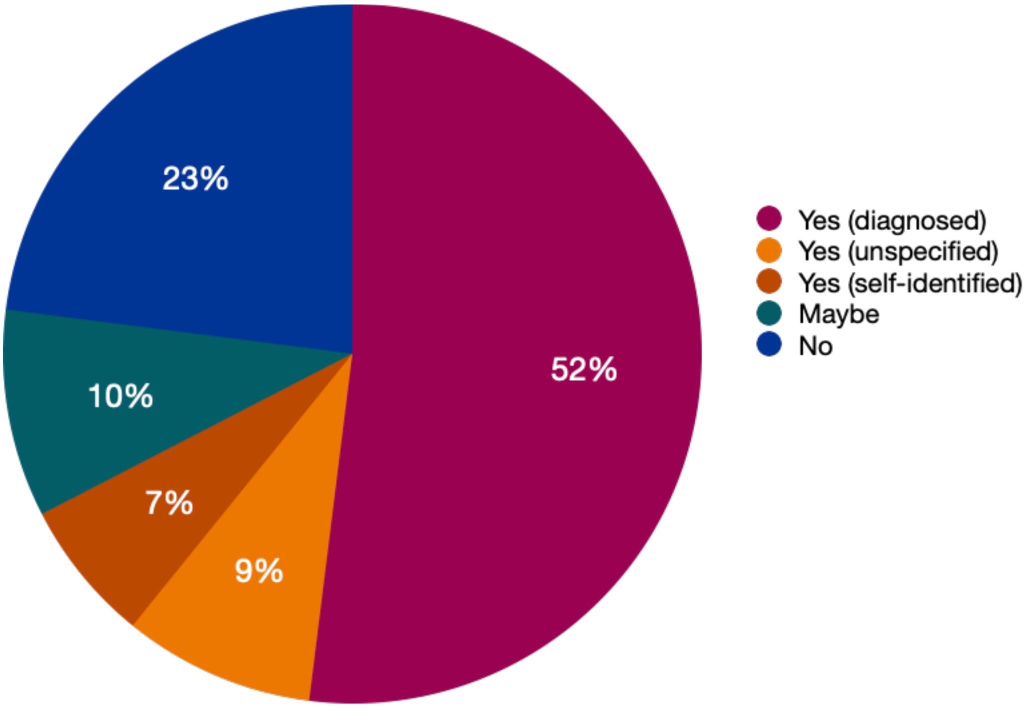
We have used qualitative research methods to collect and analyse responses to the consultation. We also used some quantitative methods to support this. To enable autistic people who have difficulty expressing their ideas in words to make their views known, we have provided some questions where consultees can endorse the option that is closest to their view. Transcripts were created from oral and text interactions and combined with text responses from the survey. These were anonymised so that no one can be traced back to the comments they made in this report. We call the people who responded in Phase 3 ‘consultees ’ throughout this report.

In qualitative research we do not report on the number of times something was said, but rather the strength of feeling expressed across the methods used. We use grounded theory which means we read, and re-read, the transcripts many times. We collate what was said into key themes and from these draw out the meaning from the discussions.

1.8. Who responded to the consultation?

The consultation was publicised on the Hopkins Van Mil and Spectrum 10K websites, on social media, and by email to those who had expressed an interest. In all, 531 individual responses were received 457 to the standard survey and 74 to the Easy Read version).[8] 358 of these responses were from people who identified as autistic, and a further 51 who thought they may be autistic.[9] 122 people who responded to the survey are not autistic but described themselves as parents, carers, partners or other family members of someone who is autistic. As illustrated in Figure 4, 67% of respondents across both surveys identified as autistic. In total, 55 people attended the online webinars and discussions. Consultees were drawn from a range of demographic backgrounds, including people who identify as male, female, or non-binary, people of different ethnicities and ages, and people with different kinds of co-occurring conditions and disabilities.

Figure 4: Answers to the question "Are you autistic?"



Note: The Easy Read version of the survey did not ask consultees to specify whether they had a formal diagnosis of autism, so these are unspecified.

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[8] The total number of interactions with the consultation surveys was 1041 (891 with the standard survey and 150 with the easy read). Interactions were only classed as a response if they came from a unique IP address and went beyond initial background questions.

[9] This number includes people who responded with either 'Yes, with a formal diagnosis' or Yes, not diagnosed' to the survey question 'Are you autistic?' in the standard survey or 'Yes' to the same question in the easy read survey.

1.9. A note about this report

This report explains what we heard from those who took part in the consultation, the ‘consultees’. Their comments represent their thoughts, concerns, fears and hopes for Spectrum 10K. The majority of those who took part are autistic (see Figure 4). The report has been written by the HVM team and Leneh Buckle.

Throughout the report:

- Bullet points are used to summarise key points made, these mostly reflect areas of agreement and where points were made by many people across many of the groups
- Terms such as ‘a few’, ‘several’, ‘some’ or ‘many’ are used to reflect particular areas of agreement and difference
- Anonymised quotations are used to highlight points made by a number of consultees. These quotations are not edited so as not to distort the speaker’s meaning.

2. Autism community and relationship with researchers

The consultation was initiated in response to vocal opposition to the study. This section gives some of the background to that opposition, which represents some of the strongest fears about genetic research and its implications. These fears are rational in the context of historical efforts to eradicate specific conditions, or autism in particular, but they are not always related to Spectrum 10K specifically.

2.1. History and track record of research team

Opinions about the research team within the autistic community are polarised and passionate. While some of the research team have a long and prominent history in autism research and are well known and admired in the field, negative associations with members of the team or with genetic research means some consultees immediately mistrusted the study.

> *Autistic people cannot trust the researchers involved regarding this study, therefore it is completely impossible for the autistic community to be fully confident that team members no longer hold these harmful views about autism.*

Some consultees question the motivations of the researchers and funders. Although the Spectrum 10K team have stated that they do not wish to cure or eradicate autism;[10] some have so little trust in the research team that they cannot accept anything they say, leaving no scope for clarification.

Composition of the research team

Specific concerns have been raised about Dr Daniel Geschwind due to a past association with the now dissolved organisation Cure Autism Now.

> *While Daniel Geschwind remains involved with this project, confidence will be damaged as it cannot be 100% proven that he no longer holds the view that autism can/should be cured.*

Many consultees believe that having autistic researchers on the team is essential for both ethical and practical reasons, and that the study should not continue without openly autistic researchers involved.

> *The only way to get decent data from autistic people is to have autistic people lead the study.*

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[10] https://www.newscientist.com/article/2179104-genetic-studies-intend-to-help-people-with-autism-not-wipe-them-out/

2.2. Public objection to Spectrum 10K and response

When the Spectrum 10K study was launched the team were unaware of the extent of suspicion that the team had a hidden eugenics agenda. As described in the introduction this led to a mixed reaction to the study, some showing immediate support and some opposition, including a boycott campaign.[11] Some of the main concerns are illustrated in the following quote:

> *This study is beyond saving. Like in the previous answer: in a way which doesn't do justice to the many problems with Spectrum 10K – so many issues remain around personal data, the previous work of researchers, the potential commercialisation of genetics data, the lack of involvement of autistic people and more, that the study is completely unworkable and unethical, and must be shut down.*

Much of the reaction took place on social media, but also included letters, blogs and other articles.

For some consultees, Spectrum 10K's response to those who have called for the study to stop entirely has been problematic. There are two main contrasting reasons for this - a belief that Spectrum 10K:

1. Responded in a way that has been seen by some consultees to show a lack of sensitivity and empathy.

> *Spectrum 10K has interacted with the autistic community online in a dismissive - and sometimes even combative - way that has resulted in the depletion of any trust that once existed that the organisation genuinely has the best interests of autistic people in mind when making decisions regarding the proposed study.*

2. Did not do enough to address criticisms and allay fears.

> *I think a lot of misunderstandings have occurred. I think Spectrum 10K should have been more active on social media to explain and answer questions.*

Many of those who believe the study should continue fall into this second category. They believe that more could have been done to transparently address concerns swiftly so that the study could have continued sooner.

> *The study can be improved by not pandering to these shouty people. The research is important and most of us could see that it was not going to*

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[11] https://aucademy.co.uk/2021/09/03/boycott-spectrum-10k-please-sign/

> *be for eugenics or whatever ridiculous things the online people said. Please start the study again.*

2.3. Communication difficulties

Some objections to the study arise from clear misunderstandings, such as a belief that there are commercial motives or that there is collaboration with American organisations. These misunderstandings indicate a need for clearer communication about the funding source, collaborations, and future applications of findings from Spectrum 10K.

Some communication failures are more complex. One example, of many, of a breakdown in communication is given here. During the development of the consultation (see Phase 2 report) concern was raised about how inaccuracies in medical records may affect study data. The research team sought to allay those fears by saying that, by using multiple sources of data, 'Spectrum 10K can take into account possible inaccuracy in medical records'. This has then been interpreted as the researchers setting out to locate inaccuracies in medical records on an individual basis, as illustrated by the following quote:

> *It is completely inappropriate and unethical that a research team might identify discrepancies in a participant's health records before they do, and/or not inform them of such errors.*

Furthermore, the use of multiple sources of data to mitigate the effects of inaccuracies has been taken as evidence of mistrusting autistic people.

> *If medical info is being given in questionnaires, the only need for medical records would be because you don't believe people's self-reports of their issues, then either the questionnaires or records are useless.*

For some consultees the lack of trust is too severe for any clarification to be trusted; however, for others, clarification of points of misunderstanding or confusion may help.

2.4. The problem of representation

The autism community is not a united group with a single philosophy. While the consultation explicitly seeks the voices of autistic people, consultees say no one person represents 'what the autism community thinks' or what 'autistic people want'.

> *If you must include more autistic people make sure they ARE diverse and not just the ones from the echo chambers on social media! A lot of us simply do not care about puzzle pieces and fighting over terminology, but if you look on these groups you would assume they speak for all of us.*

Some consultees are concerned about divisions within the autistic community. They believe that the study should do all it can to be inclusive of all autistic people whatever their situation or needs. This includes non-speaking autistic people who are likely to use other written/ visual forms of communication. There is also a concern that some groups within the autism community are shunned by others, for example non-autistic parents of autistic children, or autistic people who want to see a cure or treatment for autism.

> *For many parents, including those like me who are official Deputies for our adult sons/daughters, our voices are the best way of getting the viewpoints of our children. We are 24/7 carers, why are we so often treated as chopped liver?*

Some consultees suggested non-autistic parents may have an unrealistically negative view of autism and are concerned that the views of parents are considered equal to those of autistic people.

> *Parents who are not known to be autistic can have ableist views of autism and may not fully understand or appreciate the ethical concerns with the study. In day-to-day life parents of autistic children often come across fear mongering which may make them feel compelled to look for answers for their child which in actual fact are not helpful nor ethical.*

Some consultees point out that people who can speak for themselves are only part of the autism community.

> *Spectrum10K needs to make a decision as to what is best for ALL people with autism, not just those who are verbal and able to communicate, and to pursue its aims without prejudice to a certain vociferous group.*

Other consultees point out that even ‘those who are verbal and able to communicate’ do not share a single view.

> *I often feel frustrated when I hear autistic people speaking on behalf of all autistic people when I do not find what they are saying is relatable to my own experience at all.*

There is a strong feeling within responses to the consultation that those who object to the study may fail to consider the views of those whose experiences are more negative or mixed.

> *I agree that autistic people need to be involved in autism research. I am also concerned that the slightly militant branch of autism-positive people are dismissing the experiences of autistic people who are unhappy as they*

> *are, or those who are less able to voice their experiences. Not everyone is ok with being Autistic. Some are tortured by it.*

2.5. Support for the study and scope for improvement

Consultees have a range of opinions about genetic research in general and Spectrum 10K in particular. While all views were represented in all groups, people who did not identify as autistic, such as parents or carers of autistic children, are more likely to have a positive opinion about the study. There are several themes within the views expressed, ranging from a call to stop the study immediately to a plea to start it again urgently.

The only way forward is to stop the study

For some consultees, a fundamental disagreement with the pursuit of any genetic research for autism prevents acceptance of the study, regardless of who conducts it or how it is modified.

> *It cannot be improved because the way the study was designed laid the foundations for a dangerous study. Because "One of the terms of the funding agreement for Spectrum 10K is that there must be a genetic element to the study", you can't remove the most concerning element of the proposed research.*

In stark contrast to this, there are those who are wholeheartedly in favour of the study and do not feel it needed to be paused or amended.

> *I strongly support research into Autism and I wish more research focused on the biological and medical aspects. I am annoyed that Spectrum 10K has been paused for so long.*

Scope for improvement

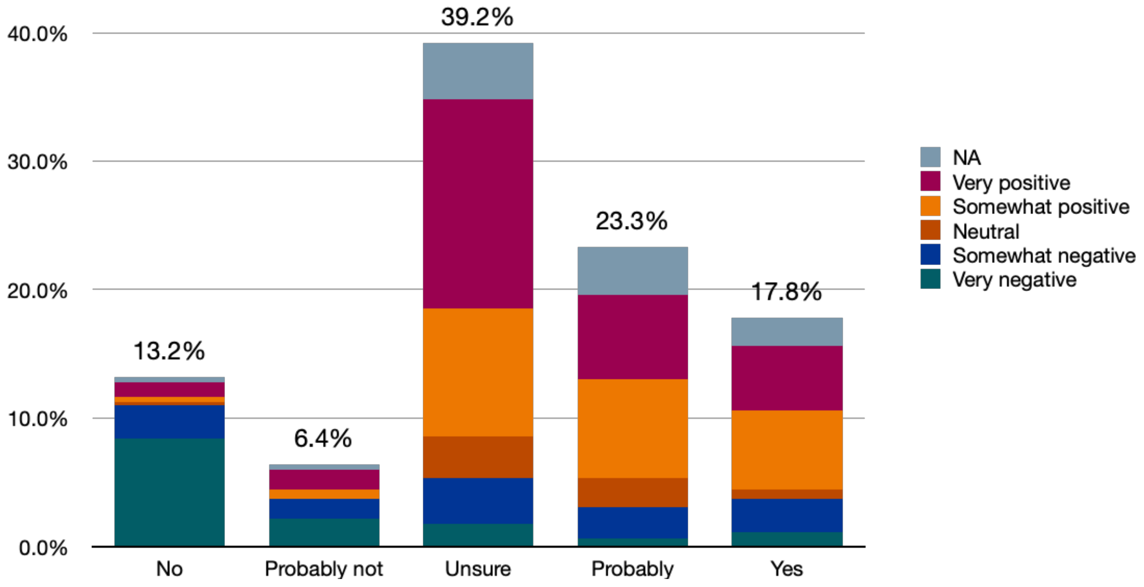
Many consultees had a mixed opinion about the study. They are not categorically against all genetic research, but have some reservations about Spectrum 10K. Because the purpose of the consultation is to improve the study, consultees were asked whether they thought the study could be improved. A majority of consultees responded that they believed there may be scope for improvement. (See Figure 5 for details.)

Some consultees welcome the consultation as an opportunity to amend previous errors or oversights, but still see value in the study as it was originally conceived.

> *Absences from the original design can be dealt with by a later involvement exactly like this one. It would be fanaticism, and discrimination against non-autistic workers' thinking capacity, to completely throw out an idea because who thought it up differs to a perfect demographic ideal!*

It is with this background understanding that we approach reporting the results of the consultation. Within each topic area there are those who responded simply that the study should be stopped. It is useful to acknowledge this and then to look at the more substantial comments.

Figure 5: Answers to the question "Do you Do you think it is possible for Spectrum 10K to be improved?"



Note: The responses are separated by answers to the initial question about how they felt about Spectrum 10K. These are represented by the colour bands in each column.

Recommendations

- Consider the recommendations for communication detailed in Section 7. Specifically, provide clear, explicit, consistent and detailed information about:
    - The research team, their role in Spectrum 10K, and a statement about their goals and values regarding genetic research
    - The source of funding for Spectrum 10K and the likelihood of commercial application of the results,
    - The motivations of funders and researchers.
- Apologise publicly for insensitive comments.
- Respond quickly to questions from the public about the study. Delays can raise suspicions that there is something to hide.
- Consider the recommendations for involvement detailed in Section 7, in particular:
    - Consider recruiting one or more autistic researchers and/ or expert consultants.
    - Include a wide variety of autistic people and other stakeholders (e.g. family members).

  - Seek out those who bring multiple perspectives, e.g. autistic people who are also parents of autistic children, representatives of autistic people's organisations, and those who have multiple marginalised identities (e.g. ethnic minorities).

Part two: Aims and intentions of the Spectrum 10K study

3. Genetic contributions to autism

3.1. Lack of clarity about the aim to look for genetic contributions to autism

Critics following the study launch, and consultees point out the primary objective "to accelerate gene-discovery, genetic stratification, and biomarker identification" in the grant proposal for Spectrum 10K,[12] differs from publicity about the study, which focused on co-occurring conditions and wellbeing. Some consultees accept the research team's explanation that a study's aims – and the ways in which these are described – may develop over the course of a project.

> *I really like how expressive you have been in telling us that this is not about preventing autism. I believe many people that are close to someone with autism would agree with this and find it very ethical.*

For others, such inconsistencies indicate a lack of transparency about the study and have undermined confidence in the study. Many of the more critical consultees say that the aim to find genetic underpinnings of autism and autistic traits has been deliberately hidden from potential participants and the public.

> *That the Spectrum 10K team hasn't been clear from the start is indicative that they are not capable of running this study ethically, in my view.*

While some consultees do not believe it is possible for the research team to conduct this study ethically, others believe that greater clarity could increase support.

> *I think their needs to be clarity on why you want to find those things out. If you want to explore the genetics of co-occurring conditions, you have to say WHY. Lots of Autistic people are scared of others trying to "fix" them when they don't need fixing. Without knowing WHY the study wants to explore these issues, that's what they'll default to. Some of us, however, would desperately like to have those areas explored because, God knows, we don't want to live with the mental health issues anymore.*

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[12] Many of these were released through multiple Freedom of Information requests as detailed in this personal blog: https://gizmonaut.net/blog/autism/2022/11/spectrum10k-grant-application.html

> *Without clarity, no one can take an informed stance on the purpose of the study. It's like saying you want "applied for the spade" so you could "dig the hole" without saying what the hole is for.*

A few consultees approve of the aim to develop more basic understanding of autism.

> *I think it's a brilliant idea and long overdue.*

Others feel that Spectrum 10K's aims are not based on the priorities of autistic people and their families.

> *It's a shame that they didn't consult autistic people about the aims of the study prior to it being funded. Adapting the aims to include other areas because it's clear that the existing aims were not supported is not enough to reduce the harm this study will cause.*

Seeking to understand the genetic causes of autism and co-occurring conditions are considered to be benefits by some consultees and potential sources of harm by others.

> *I'm not a fan of the "trying to see what makes people autistic". The health issue part is great, but finding out what makes us autistic is a dangerous game. It may not be used for harm by this study, but you can't control what others use it for, and it could very quickly become used in eugenics.*

3.2. The risk of eugenics

Is Spectrum 10K aiming to develop a prenatal test?

One of the main issues raised about Spectrum 10K after the study's launch, and a key area of concern during the consultation, is that the findings will be used for eugenics, such as selective termination of fetuses with a high probability of being autistic, or attempts to treat or cure autism itself. Spectrum 10K have stated that they do not aim to develop a prenatal test and that a definitive test is unlikely to be possible given the genetic complexity of autism. Still, some consultees say that even if the likelihood of a prenatal test being developed is low, the potential for harm that it represents is high.

> *As someone involved in autism research, I understand that a prenatal test or "cure" for autism is highly unlikely - but not impossible.*

Some consultees for whom prenatal testing has been a concern are reassured by the comments Spectrum 10K have made during the consultation.

> *Assuming that Spectrum 10K is being honest about their intentions, I am optimistic that the trust of the [participants] will not be abused and that the data will be used only for the purposes outlined.*

As discussed in Section 2, some consultees remain unwilling to assume that the research team are being honest about their intentions, thus no reassurance from the research team will affect their view.[13] Many consider the development of a prenatal test to be an inevitable outcome of genetic research.

> *I think just stating you would not support prenatal testing is an empty statement as it's what will happen if you find the cause.*

Because of this, for some consultees the only satisfactory response is not to go ahead with the study.[14]

Could the results ever be used to prevent the birth of autistic babies? (810)

One view frequently expressed is that the team have not adequately considered that the study could be a stepping-stone towards the development of prenatal test for autism by others, even if it has no intention of developing one itself. This concern is felt both by those who are generally supportive of Spectrum 10K and by those who oppose it.

> *It's clear but you HAVE TO ACCEPT that your research could end up playing a fundamental role in the eventual development of damaging and harmful prenatal tests.*

Views about this topic often place Spectrum 10K within a wider context. It is not only how Spectrum 10K investigates autism, but how its findings could be used by others. It is also not only about how people working on Spectrum 10K view autism but also broader societal attitudes.

> *Spectrum 10Ks position will not lead to harm, however, can spectrum 10K guarantee that their results will never be used by someone else for prenatal testing. I think not, and I think this is the crux of the problem for me and a lot of other autistic individuals. It's not what YOU do with your data, it's what someone else might do with it. Is that reason enough not to do it?*

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[13] Although it is not related to Spectrum 10K, thus outside the remit of this consultation, several consultees raised concerns related to a study run by the Autism Research Centre involving amniocentesis. We would encourage Spectrum 10K to make a clear statement to clarify the purpose of that study and any connection with Spectrum 10K.

[14] A few consultees would support prevention or treatment for autism, but this is outside the aims of Spectrum 10K, thus the remit of the consultation.

While some consultees consider these issues to be enough to stop the study, others consider how Spectrum 10K could continue whilst responding to this concern. They would like to see Spectrum 10K give more consideration to the possible consequences of its findings beyond the study itself and to be honest and transparent about this. It is not adequate to say that Spectrum 10K is not looking for a prenatal test for autism. The actual risk, however small, of the study's findings contributing to a prenatal test and selective termination, needs to be clearly acknowledged.

> You might not be developing a prenatal test but how can you prevent other people developing your findings into such a test? This question seems to be ignored each time it is asked.

The team are also urged to acknowledge the risks that come from publication of the GWAS (Genome Wide Association Study) data and the derivation from this of polygenic risk scores, which are core aims of Spectrum 10K, which could in principle be used for prenatal selection by other scientists and clinicians.

Many consultees state that it must be clear now and in the future that data cannot, and will never, be used for certain purposes including treating or eradicating autism. The recommendation is that the team state this risk in their publications and elsewhere as a warning to other researchers that such an application of their data would be against the values of the team who collected the data.

Some consultees' fears about misuse of the data come from the lack of specific data sharing protocol at the time the study was launched.

> I completely agree with S10K's position on prenatal testing and interventions for autistic people. However, this isn't enough for me to hand over my DNA in confidence that it will not harm autistic people. Without sufficient safeguarding in place, there is a strong risk that future researchers will use this dataset in ways that do not align with these aims.

Consultees say that the proposal of a DAC that includes autistic people could create such safeguards, as discussed in the next section.

3.3. Safe sharing of data

Allowing other researchers to access data collected by Spectrum 10K – often referred to as 'data sharing' – is an essential part of the funding agreement between Spectrum 10K and the Wellcome Trust who provide the funding. Data sharing is considered good practice in research[15] because it allows other researchers to benefit from a single data collection as well as to check the research was done correctly. Despite this, a few consultees say that no data

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[15] Alter, G., & Gonzalez, R. (2018). Responsible practices for data sharing. *American Psychologist, 73*(2), 146–156. https://doi.org/10.1037/amp0000258

access should be granted because Spectrum 10K could not protect the data sufficiently. Other consultees support the sharing of data so long as it does not attempt to "*cure our natural advantages and differences.*"

Concerns about the possible misuse of data are detailed in Section 3.2, and how this will be prevented is a key concern of consultees.

> *I think the spectrum 10Ks position on prenatal testing is clear however again it states that they have no interest in eugenics but if this data is given to other researchers scientists how do we know they don't have the same interest? Again this is not clear and it should be to reassure us. Sometimes its what is not said in the spectrum's statements, what if's etc..*

The policies and procedures that would govern data sharing were not specified at the time of the original launch of Spectrum 10K and this was a significant part of initial concerns about the study. Consultees reiterate this concern and ask for this to be corrected prior to any re-launch of the study.

> *I think the rules could be much clearer like how they know what type of research is allowed to be shared or not, who makes that decision?*

They have asked for a thorough procedure to ensure that those asking for access to the data have been thoroughly vetted for adherence to the stated aims and values of Spectrum 10K.

> *The people requesting to view and use the data that was collected from this project should have to go through rigorous testing before being allowed to access the data to ensure they won't use it for harmful purposes, like eradicating autism. They should be made to sign a legal document stating that they understand that they are prohibited from using the data for prenatal testing, finding cures, preventing and eradicating autism on any level.*

Figure 6 illustrates our recommended approach to deriving aims and data access protocols that are founded on a trusting and transparent relationship with the autism community.

Some consultees have practical concerns about the accessing of data, including whether people who have approved access are able to copy, share, publish or leak it elsewhere and how those using the data will be monitored and held accountable.

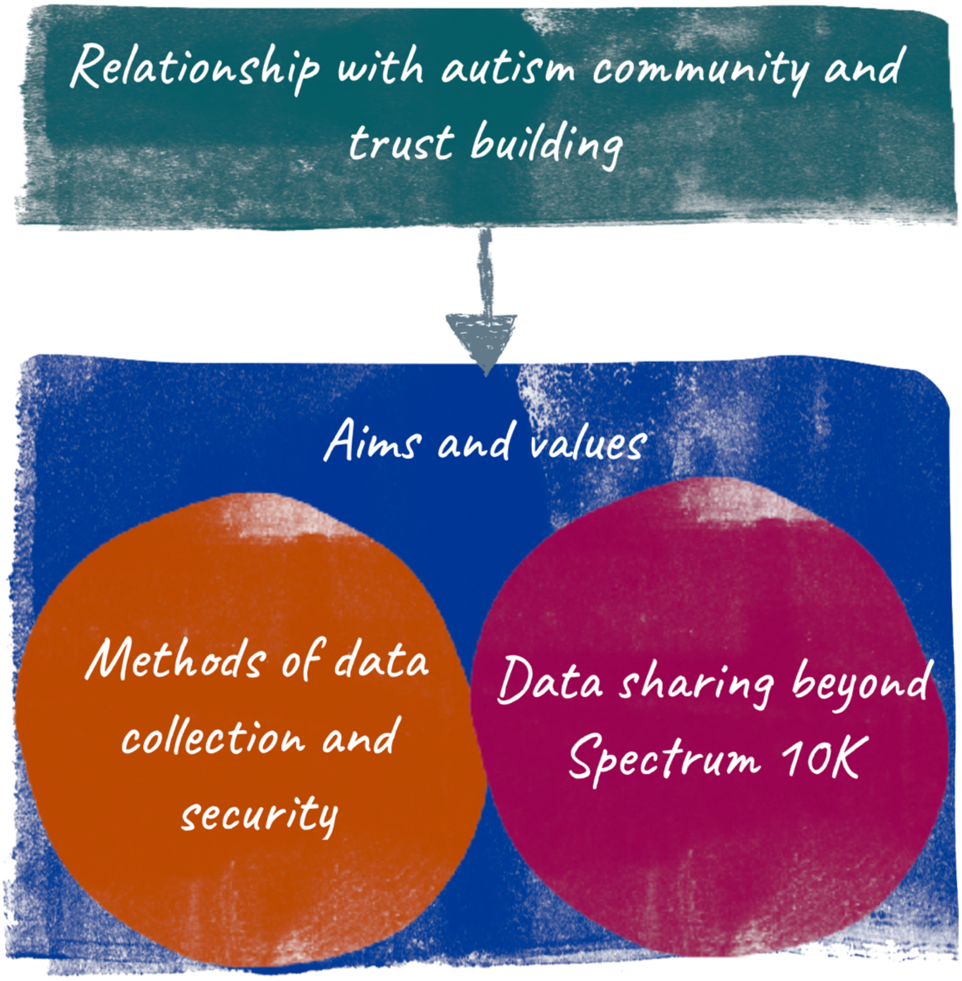
> *How will you guarantee that people accessing the data are doing so for the right reasons - People often say things to gain ethical*

> *approval/funding etc. and then change this plan. Once someone has access to the data how would you retract this? Also how would you prevent data being passed on by someone who has accessed it.*

And some have practical concerns about how the DAC will work in the longer term.

> *What is the long-term safeguarding plan? Is there possibility to change DAC membership, or add new members to replace those who have to leave? What will the process be for this? How will autistic DAC members who are not there as part of another job be fairly compensated for their time?*

Figure 6: Illustration of our recommended approach to developing trustworthy research



Note: This figure illustrates how trustworthy research begins with a firm foundation in long term engagement with the autism community. This gives rise to aims and values they have faith in, which provide the ethical background that is reflected in data collection and in data sharing.

No matter how robust the data access protocols and the DAC, some will not trust anyone else to decide what is an ethical use of their data. They want the opportunity to give consent for every situation in which the data is used.

> *Participants agreed to participate in Spectrum 10K's study, but not the third party studies. Individual participants should be contacted with each third party's objectives and allowed to decide whether their information be used or not.*

Some suggest that potential harm could be mitigated by reviewing both Spectrum 10K's own publications and those of any other studies that use the data.

> *I'm not sure that the study can of itself dictate how people in the medical and wider society use any data or conclusions and therefore it is not possible to mitigate against all bad or misguided actors. The study needs to make sure all parts of the autistic community are meaningfully involved in the production of any wording for reporting study results and conclusions.*

To many consultees, it is important that the DAC include autistic people. Some think the DAC should include only autistic people, while many think autistic people should have a controlling majority or veto power for the approval of any access request. There are divided opinions about the inclusion of non-autistic parents of autistic children, although this conflict can be avoided by seeking out parents of autistic children who are themselves autistic.

3.4. Recommendations for the Data Access Committee

Access to the data should be determined by a **Data Access Committee** (DAC). This is a key recommendation from the consultation.

Remit of the DAC

The DAC should have the following purposes:

- Reviewing data access requests for their suitability including adherence to ethical guidelines and the interests of autistic people.
- Reviewing pre-publication research that has used Spectrum 10K data.
- In cooperation with Spectrum 10K, develop specific guidelines for data access requests, which should be regularly reviewed with input from autistic experts outside the committee itself.

Composition of the DAC

- The DAC must include autistic people.
- In addition to representation by neurotype (i.e. whether one is autistic or not), also include a range of experience and expertise:

    - Being autistic isn't enough by itself. Autistic members should have relevant experience/expertise, e.g. autistic researchers, geneticists and clinicians.
    - People who have a depth of understanding of the potential harms/ risks of using the data
    - People with experience, skills and training in medical ethics
    - People experienced in protecting the legal and human rights of the wider disabled/ intersectional communities for genetic research and data release to the wider scientific community e.g., representatives of autistic-led advocacy organisations.
- Those on the DAC should share the view that Spectrum 10K data cannot be used to develop a prenatal test or with an aim to cure or eradicate autism.

Operating procedure

How the committee should work:

- It is recommended that autistic people have a controlling majority or veto.
- The DAC should encourage peer reviewed/published research using data from Spectrum 10K and review such research prior to publication.
- There should be a plan for practical issues such as payment and succession.
- Set the parameters and principles for acceptable research purposes and what can and cannot be done with the data.

Data Access Policy

There should be a clear governance structure to ensure data is held and access is managed to the highest standards with the following features in the Data Access Policy:

- Researchers' credentials should be verified and rigorously tested to ensure they have the appropriate qualifications and experience to access the data.
- Researchers should only access the data within a Trusted Research Environment (TRE), with measures in place to prevent copying the data.
- Access should only be granted to ask specific queries of the database, not to access the complete dataset. (E.g., one could ask 'Are there common genetic factors between autistic people who also have hypermobility?')
- Researchers' use of the data should be monitored over the long term.
- Protections should be in place against the 'commercialising' of genetic data.
- An experienced and specialist data controller should be in role to manage the policy.

Recommendations

- Spectrum 10K should communicate its aims openly, ensuring these are clear and consistent. Their meaning shouldn't be lost if they are summarised or adapted for different audiences.

- It is important to recognise that concern about Spectrum 10K's role in the development of a prenatal test for autism hasn't yet been fully addressed. Publicly commit to the stance against prenatal testing for autism.
- Maintain a clear stance against prenatal testing for autism and any research that points in this direction. Amplify the voices of autistic people concerned by developments around this.
- Be a positive public advocate for autism – actively communicate to increase societal awareness that autism doesn't need to be cured or prevented.
- When assessing, documenting and reporting risks, include the risks for the population being studied, i.e., for autistic people more broadly and not just for the individual participants.
- Be clear in publicity and participant information about how GWAS data that will be published can contribute to polygenic risk scores and how these may be (mis)used.
- Ensure that there is a clear ethics statement as part of any publication discouraging the use for prenatal testing or selective termination, and emphasising how little these genes contribute to autism overall.
- Make it possible for participants to consent to each future use of their data.
- Convene a DAC to review requests from other researchers for access to Spectrum 10K data. Specific recommendations for the composition, remit and policies of this committee are detailed in Section 3.5.

4. Further aims and potential benefits of the study

4.1. Potential benefits

There is widespread agreement across demographics and views about Spectrum 10K that that autistic people need better support and more understanding, but there was disagreement about whether Spectrum 10K could support this aim. They question the value of a study such as Spectrum 10K when, in its own words, "There are no direct benefits of taking part in the study."

> *If I were offered a therapy, whether medical or psychological, to make me non autistic I would not take it. I love my autistic self. Yes there are challenges but they are greatly outstripped by the benefits. I have read all the spectrum10k ethics and grant proposal and I am not convinced that this study benefits autistic people. This money would be better spent exploring what makes our lives difficult and how to mitigate these things.*

Some consultees consider that any increase in knowledge about autism will improve the ways in which people in society more generally view, understand and interact with autistic people.

> *Hopefully the findings of the study will be made available to all, and hopefully help neurotypical people understand neurodiverse people better.*

> *The more that is known and spoken about autism has to be beneficial for all those affected both directly and indirectly.*

While looking for any kind of treatment is opposed by some, and the Spectrum 10K team have said that they do not support treatment for autism itself, only for symptoms that cause distress, others welcome the possibility that Spectrum 10K could lead to better interventions and/ or support for autistic people who want it.

> *I notice some people like being Autistic as it validates their lives. They may feel threatened by anything that offers to take it away. My autism has at times been a blessing but it has also made life extremely difficult over the last 20 years and I'd like it gone or at least softened.*

Some consultees interpret unknown or uncertain outcomes from the study as a possibility that they will be beneficial:

> *I think you are right to be circumspect about the potential benefits, since you haven't collected the data or analysed it yet! However, it's clear where*

> *Spectrum 10K think the benefits are likely to be and where they are unlikely to be. I hope that, assuming the study goes ahead, the benefits will become more clear as the research progresses.*

4.2. Looking for genetic subgroups

Many consultees agree that labels such as Asperger Syndrome and "Profound Autism" are attempts, historical and recent, to categorise autistic people in a way which does not always put their interests first. Views on Spectrum 10K's aim to look for genetic subgroups are polarised. Some consultees consider genetic subgroups to continue a tradition of labelling autistic people which is flawed and discriminatory. Searching for subgroups is described as a "step backwards".

> *We are trying to move away from distinctions like high/low functioning and the Asperger's diagnosis is no longer part of the DSM. Discussions of "profound autism" recently have caused distress in the community. It's not clear how identifying subgroups is a step forward.*

While agreeing with the underlying problem, other consultees took the opposite view, that genetic subgroups could help to move away from "functioning labels" such as "high functioning", which are based on weak behavioural evidence and can lead to discriminatory practice. Some say that identifying genetic subgroups distinguishing those with different co-occurring conditions could be helpful.

> *I think further distinctions help us move away from the very narrow and negative connotations of high and low functioning.... These terms are offensive but in lieu of anything better get used all the time.*

Some consultees fear that genetic subgroups will further divide autistic people when there are benefits to having a collective sense of identity or community. Conversely, others think that, in a similar way to the autism label itself, subgroups could help an individual to find others with similar experiences.

> *Autism presents in such an individual and varied way, but shared characteristics help people feel they belong, aren't alien, have shared experiences.*

Some are sceptical about the Spectrum 10K team's statement that they do not consider one subgroup to be better than another.

> *Spectrum 10K say they don't "see one subgroup as better than another", however in previous messaging they have said that the study looks to "understand the broad heterogeneity within autism that ranges from learning difficulties through to talent", implying these two characteristics are on opposite ends of a spectrum and cannot coexist (i.e. Learning*

> *Disabilities Autistic people can't be talented). Can this study team be trusted to explore subgroups if implications like this suggest they don't view all Autistic people as equals?*

There is also concern that subgroups could lead to further discrimination or difficulty accessing support.

> *OK, this is tricky because this fascinates me as a geneticist, whether there are benefits or not. At a personal level I'd love to know about this. I do worry that subgrouping could eventually result in some autistics that fall into a group getting support and those that don't fall into a defined group not.*

There are many questions about how subgrouping works, and some evidence of misunderstanding, so this is another area that would benefit from further clarification. Some examples of these questions include:

- How many subgroups becomes too many?
- Given that some aspects of autism such as anxiety or sensory processing can vary within individuals over time, how does this affect the definition of subgroups?
- Will the data that contributes to subgroups be dimensional, so degrees of the trait are taken into account, or categorical, so it is either present or not?
- Where genetic associations are found, will they be able to indicate how much influence is from the identified gene(s)?

4.3. Investigation of co-occurring conditions

While research into co-occurring conditions met with broad approval, some consultees suggest there are more effective ways to approach understanding and support for these conditions than genetic research. They want Spectrum 10K to consider the non-genetic factors that influence an autistic person's experience, such as the environment and "living in a neurotypical world". Some worry that genetic subgroups may be used to fix people within a particular diagnosis or reduce someone's experience of autism to its genetic background. Others suggest similar research could be done better – and safer – without involving genetics at all.

> *Exactly the same alleviatory medical research can be done with symptomatic subgroups. That would not carry the genetic discrimination concerns. So there is no need to bring genetics into this. It is ethically safer not to. You can still use familial relations to look at the subgroups' heritability, just without the DNA. The subgroups examples, language*

> *delay and learning disability, are defined by symptom so they already are symptomatic subgroups not genetic.*

Some consultees strongly support the aim to look for genetic subgroups and they often share Spectrum 10K's rationale for doing so.

> *I suffer with permanent gut pain - if that turns out to have a genetic subgroup associated with it then I and millions of other autistics could be cured or live pain free. These are compelling arguments that make me support genetic subgroup research.*

The idea appeals to some people because it places more emphasis on how experiences of autism can vary from one person to another.

> *I thought the aims were pretty clear. They wanted to investigate autism in the context of other medical conditions to see what role those other conditions had. This is something I was very interested in. I don't just have Autism, I also have [other conditions]. Teasing apart which condition causes which symptom is really hard sometimes and mostly I think a bit pointless. I was very much hoping that this study could help in finding the underlying explanation for people like me.*

Recommendations

- Avoid unhelpful labels. Recognise the history of labelling and grouping autistic people and the harms this has caused in the past.
- Clarify what is meant by subgroups, how they will be determined, and how they differ from previous approaches to subgrouping within the autistic spectrum.
- Make it possible for study participants to opt-in separately for subgrouping research.
- Continue to consult autistic people about the design and implications of subgrouping.
- Words such as 'treatment' should be used carefully and explicitly in connection with the aspects of autistic people's lives that they are interested in receiving treatment for (e.g., specific co-occurring conditions).
- Be clear about what benefit may come from subgroups. Ensure that such descriptions are clear, balanced and provided before consent is obtained. Specific information that would be valuable includes:
    - The path from this basic research with no direct benefit to possible future benefits.
    - The intended practical impact of any genetic discoveries about autism (e.g., treating co-occurring conditions, diagnosis).

    - What benefits are not likely, even when that is to the detriment of promoting the study. E.g., would it be expected to lead to social change?
    - A complete list of the co-occurring conditions the study intends to investigate should be published and easily accessible.

Part three: Study methods

5. Data collection and security

5.1. Questionnaires

Other than those who object to the study wholesale, the use of questionnaires is relatively uncontentious. Those who are positive about the use of questionnaires gave several reasons:

- Questionnaires are an easy and common way to gather information.

> *Research is about asking questions and getting answers, so the use of questionnaires is routine and well-understood by most people... If I couldn't use questionnaires I wouldn't be able to do my job properly, so of course I understand and support the need to use questionnaires in the Spectrum 10K study.*

- They could provide information that is more accurate than the data in health records.

> *I feel this is more accurate than health records. Many health records don't depict a true version of the persons feeling and views. Questionnaires allows the answers to be first hand and not firstly interpreted by a medical Professional to go into notes*

- They could help establish patterns of data.

A few consultees point to previous questionnaires which they believe lacked meaningful input from autistic people in their design.

> *Questionnaires regarding autism need to be peer reviewed by autistic researchers. The 2001 ARC's Autism Spectrum Quotient test used autistic people as subjects but not as consultants. Can we avoid this happening again?*

Questionnaires can be difficult for some autistic people to answer.[16] Some consultees shared why they think seeking input from autistic people into the questionnaire design is important:

- Enabling autistic consultants to suggest questions could add valuable insights.
- Helping to manage the risk of asking triggering questions.
- Developing meaningful explanations about the benefit of asking each question.
- Recognising that not all responses can be 'yes / no' – some may need an 'it depends' option or the option to include a free text response.

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[16] Rachael Stacey and Eilidh Cage. (2022) "Simultaneously Vague and Oddly Specific": Understanding Autistic People's Experiences of Decision Making and Research Questionnaires. *Autism in Adulthood.* ahead of print http://doi.org/10.1089/aut.2022.0039

> *Should include an option for free text too. Questionnaires do not fully capture an autistic persons' experience.*

Some consultees object to the study itself, thus to any data collection. This view was outside the scope of the consultation. Others suggest alternative methods of data collection including face to face interviews, focus groups and public events.

Consultees ask further specific questions about the design and use of questionnaires:

- What are the limitations of these questionnaires?
- Who is writing the questionnaire?
- Why are there questions about alcohol use and inappropriate sexual behaviour?
- Will there be questions about the positive aspects of autism such as talents related to autistic traits and life successes?

5.2. Accessing medical records

The use of medical records was far more contentious than questionnaires. Most of the consultees who are opposed to the study in general disagreed with this element of it specifically. Some of the reasons they give are:

- A well written questionnaire should provide sufficient health data, and asking for medical records suggests the study researchers don't believe the self-reported data from questionnaires. (See chapter 2.2 for details.)

> *I have been involved in the project management of many genetic studies that have not needed to access medical records due to a well written questionnaire. It is not necessary.*

- Fears about who the data would be shared with, deliberately or accidentally.
- It is unethical to receive a large amount of medical data from vulnerable groups.
- There appears to be no clear plan for sharing the data.
- Only health care professionals should access medical records.
- It is unclear how participants will be involved in the NHS Digital Data Access Request Service.

> *Do not access medical records. In England, for example, to gain access to such records, they will need to make an application to NHS Digital's Data Access Request Service (DARS), one principle of this plan involves applicants making clear whether patient objections will be upheld. There is nothing in the current plan about whether participants can see and review either the application or data access agreement, nor has a sample application been made available so participants know the extent of the information requested.*

The incompleteness of medical records and their inaccuracies are another frequently mentioned concern for consultees. The reasons given for the unreliability of medical records include:

- Healthcare professionals not viewing autistic people as individuals.

- Poor record keeping.
- Misdiagnosis of autistic people's health conditions.
- Autistic people not using NHS services (particularly if they have been diagnosed through private health services).

A few consultees say that the medical records for under-diagnosed groups, such as women, could be particularly inaccurate and therefore could lead the study to make incorrect findings.

> *Medical records for autistic people are more inaccurate than accurate, especially for under-diagnosed groups. It is the common experience for autistic women especially to be diagnosed with every mental health condition under the sun before being diagnosed with autism. This does not mean that any prior diagnoses are correct, and will lead to erroneous links.*

Those consultees who see value in using medical records want the study to explain how it will account for inaccuracies and incompleteness. (See chapter 2.1 for further discussion on this.) There are questions about who or what the Spectrum 10K researchers will believe if they find inconsistencies between the questionnaire and the medical record. Several consultees want participants to be able to highlight missing or inaccurate data or ask that it be redacted.

> *Another consideration is that misdiagnoses may be on the medical record, as is common for neurodivergent individuals. The opportunity for the [participant] themselves to request specific information to not be included/provide relevant context may therefore be beneficial.*

Some consultees say that medical records complement the questionnaires because participants may forget some information asked for in the questionnaires.

While many say that the sensitivity of the data in medical records meant making its inclusion mandatory would discourage participation in the study. A small number of consultees say they think the records are such an important part of the study that they should be mandatory; however, most of these are pragmatic and say the choice should be based on whatever will be most useful in encouraging a wide range of people to participate.

5.3. Is my data safe and secure?

Many consultees agree that written responses make clear how vital strong data security and privacy infrastructure, expertise and processes are to trust in and credibility of the Spectrum 10K study. Most responses took the form of questions, which suggests that more detailed information is needed on how data will be kept secure and private and only used for intended purposes.

There are questions and concerns about how the data will be protected in the future, after the end of the Spectrum 10K study itself. It is unclear how long the data will be retained, and some evident assumptions that this is indefinite.

> *Not to mention, data is forever, people are not. The data from the study would remain for decades, centuries into the future, and would eventually get into the hands of researchers who want to create a prenatal test for autism.*

Consultees want to know how the team can prevent future research into prenatal testing or finding a cure for autism. This is addressed in chapter 3.4.

During the lifetime of the study, some consultees would like specific details about how much is being invested in the latest security measures and the skills of data security staff working on the study.

The ability to identify an individual just from their health data was an issue that needs addressing for some consultees.

> *People can be identifiable from medical data alone without any specific identifiers like name/address included.*

Other questions raised by consultees about data security and privacy are:

- How will the data from questionnaires be stored and protected?
- What would happen if the study server was hacked?
- Are the servers dedicated to just S10K data or are they shared with other departments/schools in the university? If they're shared, what security is used to keep the data separated from other research teams?
- How will S10K ensure that all its data is securely held on the University of Cambridge's Secure Data Hosting Service, and not stored elsewhere either accidentally or otherwise (for example, through human error)?
- What assurance can S10K provide that the hosted data will in fact be secure from hostile actors who would, for example, benefit from stealing or restricting access to the data?
- What does ISO 27001 actually mean for the security of participant data?

Questions about processes:

- On what basis will researchers be given access to data? More explanation is needed.
- How will detailed health data be separated from identifying data?
- How will S10K ensure that all its processes involved with handling data are followed?
- Has the University of Cambridge's Secure Data Hosting Service ever been audited for cyber security (e.g. through penetration testing or other methods)?
- How will S10K safeguard data after the end of the project? How will they keep the data from getting into the hands of people seeking to cure autism?
- Can there be encrypted computer links between the DNA data and medical records and if so, why does a data manager need to see the data?
- Will S10K contact the National Cyber Security Centre?

Questions about GDPR:

- How will you meet the GDPR requirements for safe storage, update and deletion once the use has been fulfilled, particularly if future use is expected?

- How does S10K's long-term use of personal data meet the 'legitimate interests' requirement of the GDPR?
- How does GDPR apply, now the UK has left the EU?

Data use outside the UK

- What data could go abroad to study partners? How are the US-based researchers are going to be involved in accessing data?

One consultee expressed confidence in ISO certification of the data hosting service, but this confidence was based on professional knowledge.

> *Data security happens to be something I'm interested in professionally. It is good that Spectrum 10K's data hosting service will be ISO 27001 certified. However, this does not, and never will, guarantee that the data will be one hundred percent secure (i.e. fully secure). That said, an ISO 27001 certified system is one of the most secure systems possible and it is one of the closest things to 'fully secure' that humans can currently create.*
>
> *However, there is sometimes a difference between being accredited for something and following best practice.*

Those consultees who oppose the study, say nothing will ever guarantee data security and privacy, so the study should not proceed.

> *Regardless of who they choose to share or not share genetic data with, its very existence caused the potential for harm, which is a risk not worth taking.*

Recommendations

- Study questionnaires should be designed in partnership with autistic people to take account of:
  - Including 'it depends' or free text responses where appropriate.
  - Questions that could trigger distress.
  - Stating clearly why the questions are being asked and what benefits the answers will bring.
  - Making the questionnaires accessible to as wide a range of autistic people as possible.
- Make as many questionnaires as possible optional.
- Explore alternative channels to supplying questionnaire information if questionnaires are not accessible/appropriate for some people.
- Explain how discrepancies between questionnaires and medical records are handled.
- Allow participants to view the data held on them in the Spectrum 10K study and add to / amend it.

- Explain clearly which parts of medical records will and will not be accessed and why, make accessing medical records optional, and, if possible, allow participants to specify which parts of their record may be accessed.
- Provide clear detailed information about data storage and security. Set out more clearly how data will be prevented from being accessed by researchers who are aiming to cure autism or find a prenatal genetic test.
- Be clear about what data is collected, how it is organised, how it is stored, how it will be accessed, how long it will be kept, and what will happen afterwards.
- Create a dynamic Q&A Document in partnership with autistic people that provides clear and unambiguous responses to questions about the methodology of the study and data security, storage and privacy. Updated as questions surface.

6. Consent and withdrawal

6.1. Inclusion of adults without capacity to consent

When the study was initially designed and launched, there was an intention to include adults who lack capacity to give informed consent. Consent for these individuals would be obtained from a family member or relative who cared for the person. The consultee would perform capacity assessment before providing consent using the British Medical Association Capacity Assessment Tool on the Spectrum 10K website. If the individual was deemed to have capacity, then the consultee would not provide consent and the individual would register independently.

Involvement of individuals who cannot consent for themselves prompted criticism from some community members due to particular sensitivities around the collection and analysis of DNA. In the development of the consultation, questions were asked about whether it is legitimate for a friend, a relative, or a paid/ unpaid carer to provide consent to participate in the study on behalf of someone who has been deemed unable to weigh up its possible consequences. There were also concerns about how capacity would be determined.

There is no clear consensus amongst consultees about the inclusion of adults without the capacity to consent in the study. Spectrum 10K have stated that they are in consultation with the Health Research Authority (HRA) on this matter and will provide an update in due course.

In the consultation three main opinions towards the inclusion of adults without the capacity to consent are expressed by consultees:

1. **Include adults without the capacity to consent**
   For some it is important to include autistic adults who cannot consent because without them a substantial proportion of the autistic population would be missing. These groups are already underrepresented in research[17] in part due to difficulties obtaining consent. If the study does not include them the applicability of any results will be limited.

   > It is not possible to research "autism" without including all different kinds of autistic people. Some autistic people are not able to consent for various reasons and they need to be included in research otherwise the research will be meaningless.

   Proponents for inclusion of adults without capacity see no ethical or practical reason for non-inclusion because the procedure to take the DNA sample is non-invasive and the data is anonymised once entered into the system. They feel that this group of people may gain the most benefit from the research findings and as such are key participants for the study.

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[17] Jack, A & Pelphrey, K A (2017). Annual Research Review: Understudied populations within the autism spectrum - current trends and future directions in neuroimaging research. *The Journal of Child Psychology and Psychiatry.* https://doi.org/10.1111/jcpp.12687

Given the totally non-invasive nature of the sample-taking, and the anonymity of data once entered into the study, I don't see any demonstration of harm to these participants, so I accept the ability of someone else to essentially provide consent for them. As with children, if you rule out any Autistic adult who lacks capacity to give consent, then you end up with a self-selecting group that will skew results.

2. **Include adults without the capacity to consent – but seriously consider the ethical implications**
   Others also believe it is important to include this group of adults in the study; however, they believe that there is an ethical concern about doing so which needs to be considered before inclusion in the study takes place.

   I think it's important to include these people, as otherwise there will be a huge gap in the study, as they may have various genetic similarities that render them unable to consent themselves, and therefore their healthcare needs may not be met by analysis of the data that is gathered from other subgroups. But I do also acknowledge that it's inherently ethically ambiguous to do so.

3. **Do not under any circumstances include adults without the capacity to consent**
   Some consultees believe it is entirely unacceptable to include those who cannot consent in the study. They believe that doing so treats those without the capacity to consent as 'test subjects' with no autonomy. For these consultees inclusion is not, and never can be ethical.

   As an autistic person and a carer of an autistic person without capacity to consent, I would never enrol them to take part in something for which they could not consent unless it was absolutely necessary (life saving or to significantly improve quality of life). This study does not fall into this category.

6.2. Inclusion of children

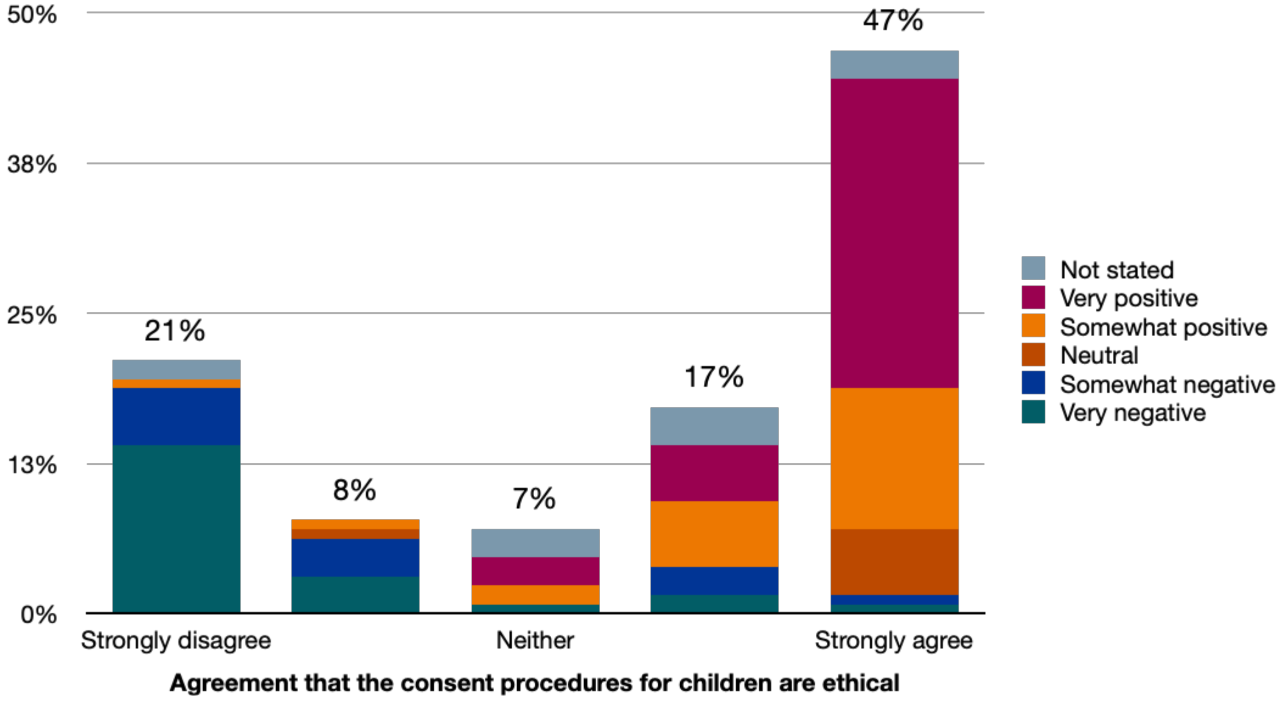
Consultees express a range of opinions on the inclusion of children as participants in the study. Comments focused on three main topics which are shared in this section of the report:

- Questions around the practicalities of re-contacting participants when they reach age 16, and the importance of obtaining informed consent at that time
- The view that children must be involved in the discussion to be included, and so can only be included after the age of 6 when they can give assent.
- The view that the inclusion of children is essential to the study.

As with adults without capacity to consent (see Section 6.1), there are concerns about involving children in the Spectrum 10K study, with the additional complication that many children will be considered able to consent at an older age. They have asked whether it is legitimate for parents or guardians to consent to taking part in the study on behalf of a child.

The existing procedures allow those over 16 to consent for themselves. For children under 6, their parents may consent on their behalf without asking for assent. For children aged 6-15, they should be provided with a child-friendly explanation of the study aims and procedure and asked for their agreement to participate, called *assent.* Most consultees think the consent procedure for children is ethical (figure 7). Most of those who strongly disagree are negative about the study overall.

Figure 7: Answer to "I think the way Spectrum 10K is handling the consent procedure for children is ethical."



Some consultees believe that young people under the age of 16 should not be included in the study as they will not be able to fully understand all the implications of sharing their DNA sample and give informed assent.

> *The project should not involve young people under the age of 16 in any circumstance as they may not be aware of the potential harm that their data could help with (e.g. prenatal testing and eradication of autistic people).*

There are specific concerns at the age thresholds of 6 and 16, and some questions about how these will be handled. Some consultees are concerned about parental pressure to be involved in the study, even if the child does not wish to do so.

> *What happens if a child turns 16 and is opposed to their involvement in the study. If a child is not assenting but parent has given consent, what would you do to ensure parental pressure does not mean their child remains involved against their will.*

On a practical level, the concern that it will not always be possible to trace the families of children when they turn 16 as they may have changed their contact details is enough for some to oppose the inclusion of children.

Many consultees believe that including children is essential to achieve the level of understanding of autism that the study hopes for. Children who may be unable to give their assent are underrepresented in autism research.[18] Some consultees also believe that non-autistic parents making the decision to include their autistic children in the study makes the study more robust. They feel it prevents the cohort being entirely made up of adults who are interested in the study and genetic research.

> *If you only use adults, it's a self-selecting group which could skew the results. Inclusion of children, where non-autistic parents may be involved in the decision to participate, I think is protective of the integrity of the study.*

Some consultees point to a lack of clarity about withdrawal and data that has already been analysed. This would apply to children who wish to withdraw on turning 16 in the same way as it does to anyone who withdraws, which is discussed in Section 6.4.

Including children in the study would be more ethical if…

Some consultees have made proposals on how to make the approach to including children as participants in the study more ethical. These include:

- Having safeguards in place so that a child cannot be forced to participate against their will.
- Make sure the information for children is a clear as possible, and includes information on why genetic data is valuable to the study, and what the risks are in sharing that information, including future researchers using the data.
- Clear, informed assent should be required for all children over the age of 6, children who cannot assent should not be included until they can assent.
- Make every effort to contact a child directly on turning 16. Those whose consent cannot be obtained at that point should be withdrawn.
- Taking care that children really are providing assent and not simply responding positively to please an authority figure.
- Ensure that the system is not dependent on a written form which may be a barrier for children.

> *It is entirely unethical to require the child to read the information and to assent via a written form. This is not inclusive nor accessible. All participants (adult and child) should be given information in a language and format they prefer, and should be able to consent or assent using their preferred form of communication also. I feel very strongly about this.*

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[18] Stedman, A., Taylor, B., Erard, M. et al. (2019). Are Children Severely Affected by Autism Spectrum Disorder Underrepresented in Treatment Studies? An Analysis of the Literature. *Journal of Autism and Developmental Disorders.* 49, 1378–1390. https://doi.org/10.1007/s10803-018-3844-y

6.3. Providing consent on behalf of someone else

A few consultees express concern around the validity of a non-autistic person providing proxy consent on behalf of an autistic child or an adult without capacity.[19]

> *However, without first hand experience carers may also lack the ability to make informed consent on behalf of an autistic person. In my opinion, first hand experience of being autistic is required to give informed consent to a study like this.*

Some consultees do not believe that non-autistic parents of autistic children can make an objective decision. They feel that it is likely that these parents will be seeking a cure or treatment for their autistic child.

> *Parents who are not known to be autistic can have ableist views of autism and may not fully understand or appreciate the ethical concerns with the study. In day-to-day life parents of autistic children often come across fear mongering which may make them feel compelled to look for answers for their child which in actual fact are not helpful nor ethical. Therefore having consent given on behalf of autistic people by non-autistic people is a concern, especially as autistic children and their families are vulnerable. Children do not have the capacity to understand the implications of genetic research.*

For child participants, one consultee wanted to see both parent/carer consent and medical ethics advisers involved as an impartial agent to challenge the research team if necessary.

6.4. Withdrawing from the study

Spectrum 10K proposed to the consultees simplifying their three levels of withdrawal to just the highest level. Consultees who comment on this proposal are divided on the issue.

> *The new plan is more ethical because it is simpler and clearer for participants and researchers to manage.*

> *It was clear enough to me when I signed up. Stick with the three levels.*

Those that support simplification to one option see it as an easier and clearer process for participants. It lends itself to a simple, one click approach to withdrawing (as opposed to emailing the study). Those who want to retain options when withdrawing gave the following reasons:

- They felt the withdrawal options were clear when they first signed up to the study.

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[19] National Autistic Taskforce (2019). *An independent guide to quality care for autistic people.* p. 19. Available from: https://nationalautistictaskforce.org.uk/wp-content/uploads/RC791_NAT_Guide_to_Quality_Online.pdf

- It is important to give people a choice of options for what type of withdrawal suits them, for example some participants may not want to hear from the study in the future, but still be happy to have their data used.

> *If continued use of previously supplied data is of value to the program then simply asking to be removed from 'contact' should still be included - some people might simply be too busy to continue involvement but still want to help the program.*

Consultees also shared suggestions for how the study's withdrawal process should operate:

- Remind people at regular points during the lifetime of the study that they can withdraw.
- Give a 'one click' option for withdrawing to make it easy for all, particularly those who would be unable to write an email.
- For those who have requested withdrawal, explain how their data has already been used and why it can't be withdrawn from those studies and what happens to data and samples that have been collected.

Several consultees take issue with the Spectrum 10K statement that 'it would not be possible to remove data from research that had already taken place'. Some say this is therefore not true withdrawal from the study.

Consultees raised a wide range of questions that they wanted Spectrum 10K to answer as it finalises its withdrawal process:

- Why can't data be fully withdrawn?
- Will samples be anonymised? If so how can they be traceable if people withdraw?
- What is meant by 'new research'? Does that mean it will be used for all of S10K, but then not passed on to future projects? Or does it mean there are multiple research projects within Spectrum 10K?
- How will researchers establish the equivalent of Gillick competency of minors or those whom others are making decision on behalf for accessing data?

6.5. Individual access to and control of data and consent

Consultees expressed a desire for individual control over almost every aspect of participants' involvement in the study, including:

- Subgrouping research
- Questionnaires
- Access to medical records
- Data sharing with other researchers after Spectrum 10K

Although there are significant practical hurdles, participants are likely to be reassured if they have as much control as possible about what is collected about them, how it is analysed, and how it is shared.

> *There needs to be a process enabling engagement with the participant to ensure that data derived is correct and is being interpreted properly. This is for two reasons: (i) medical records in this country are, unfortunately, unreliable, due to poor professional standards among clinicians; (ii) autistic people are likely to have suffered misdiagnosis of conditions, particularly if they have been diagnosed with autism late in life.*

Examples include:

- Making all questionnaires and access to medical records optional
- Which aspects of a medical record are accessed by Spectrum 10K (chapter 5.2)
- Options to annotate or correct information obtained from medical recod
- Which studies their data is used for (chapter 3.4)

In addition to autonomy about data collection, some consultees ask that participants are able to access the data collected about them as well as the findings of the study that apply to them.

Recommendations

- Report the outcome of discussions with the HRA regarding the inclusion of adults without capacity to consent.
- Require assent from children over age 6 or with the ability to understand the adapted study information.
- Provide information and recording of assent in different formats for accessibility.
- Withdrawal process: consider a two option withdrawal process: No further contact and no further use.
- Explain why data cannot be removed once analysed.
- Recommendations around consent for data sharing have been made in Chapter 3 and for access to medical records in Chapter 5.

Part four: Going forward

7. Ongoing communication and engagement

7.1. "Nothing about us without us"

Many consultees believe that the continued involvement of autistic people in decisions about the study is essential. A number quote the civil and disability rights phrase, 'nothing about us without us' and some criticise the study for lack of robust and meaningful involvement at an early stage. These consultees make proposals for the inclusion of autistic people to guide and advise the researchers such as:

- Ensuring autistic researchers are part of the study team.
- Having autistic people as a majority say on the Data Access Committee.
- Involving autistic people in meaningful ways with regular discussions about how the study is progressing and what the findings mean for autistic people.
- Working with autistic people to test communication tools, e.g. the website; and to write social media / communications messages.
- Ensuring the autistic people who are involved in guidance on the study are diverse in demographics, opinions and experiences.

> *I absolutely believe that Autistic people need to be involved, but it's important that the ones who are accept the experiences of others, even if it is in contradiction to their own experiences.*

In contrast, a few consultees expressed the view that there is no need for further involvement with autistic people beyond what has been done in the consultation.

> *Autistic people were consulted at an appropriate point (after funding was secured) and I feel like we have been given good opportunities, including this one, to have our say.*

'Too little, too late'

A few consultees remain concerned that the study did not involve enough autistic people prior to the submission of the funding application. They express anger and frustration that the Advisory Panel only included three autistic adults. They feel that the design of the study without more substantial involvement of autistic people was wrong, and that it is now too late to put this right. These consultees express the view that the consultation has been paying lip service to involvement.

> *There is no point trying to bolt-on autistic involvement in this project at this late stage. The project's main aims are already set and funding secured based on this. Adding on a few decorative gestures at this stage does nothing to address the fundamental lack of autistic involvement in this project from day one. Disgraceful.*

Others welcome the consultation as an opportunity to amend previous errors or oversights, but still see value in the study as it was originally conceived.

> *Absences from the original design can be dealt with by a later involvement exactly like this one. It would be fanaticism, and discrimination against non-autistic workers' thinking capacity, to completely throw out an idea because who thought it up differs to a perfect demographic ideal!*

Engagement opportunities

Consultees who support ongoing involvement believe that opportunities should be widely publicised. They call for these opportunities to be clearly explained on the Spectrum 10K website and in social media. Explanations should include the details of what is required, how long the involvement will last and what the expected outcomes are. The co-designers of the consultation expressed the view that autistic people should be at the heart of the consultation. This opinion extends to the study with consultees saying that the voices of autistic people should hold greater weight than those of non-autistic people.

> *Autistic people should be at the heart of this project. There are autistic people in all walks of life, including researchers, doctors etc and these people should be at the head of any research. Whilst it would be beneficial to include neurotypical parents of autistic children to have their viewpoint, their voices should not be held higher than any autistic person.*

7.2. Open, ongoing communication

To avoid repetition of difficulties caused by unclear communication (see Section two), consultees express a desire for communication that is transparent, honest and accessible. A number of questions posed in the consultation have been set out in this report, and these should be answered in documentation about the study. Going forward, many consultees share a desire for there to be more information published about the study and the research findings. For example:

- Regular updates on the progress of the study and milestones achieved.

  > *When the study begins and following the data analysis, it would be good to have updates and sharing of milestone results, if that is possible prior to formal publishing of those results.*

- Published details about what further research has been done as a result of the study.
- Updates to study participants on how their data is being used and how they can be involved in further research on related topics of interest.

Some consultees also call for information to be published and shared widely to provide more positive information on autism and the challenges faced.

Many consultees specify that Spectrum 10K should use language with thought and care. They ask that in all communications words are used which recognise, affirm and validate the value and worth of autistic people. A few consultees call specifically to avoid words such as 'risk' in relation to autism.

> The use of words like "risk" in reference to autism inheritance only works to further scare the general public about autism.

Such positive and sensitive communication is essential if Spectrum 10K is to be trusted to hold the best interests of autistic people as a core value.

7.3. Enable access by as wide a range of autistic people as possible

Many consultees say the study will best achieve its aims if there are a diverse group of participants taking part. They want researchers to strive to improve the balance of representation in autism research.

> Include diverse range of individuals paying particular attention to race and gender specifically as excluded from previous studies and autism research in general. I feel autistic white males have been studied the most and others neglected.

Many consultees want to ensure that the study, and the communication about it, is accessible and inclusive. They expect study materials to be written in a variety of formats using Plain English and Easy Read as standard. Communications should also, in their view, be reviewed by autistic people to ensure it is clear and accessible.

> It would be beneficial to have autistic people of different levels of ability to review the formats of the website or tasks, so that you know if it's accessible for all autistic people taking part in the study.

Recommendations

- A number of questions posed in the consultation have been set out in this report, and these should be answered in documentation about the study.
- Communicate frequently and openly about Spectrum 10K's developments and about how the study's results have been used.
- Choose language with care: words like 'risk' and 'causes' can be taken as negative and oriented towards prevention.
- Provide clear, explicit, consistent and detailed information.
- Arrange for review of communications by autistic people with relevant expertise prior to publication.[20]
- Produce all study documentation in multiple formats to facilitate access.
- Create opportunities for ongoing involvement and consultation on this and other projects, and advertise these widely.
- Seek to include the widest possible range of views and experiences, including those who are critical of genetic research and those who are negative about being autistic.

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[20] Crompton, C. J., Ropar, D., Evans-Williams, C. V., Flynn, E. G., & Fletcher-Watson, S. (2020). Autistic peer-to-peer information transfer is highly effective. *Autism,* 24(7), 1704–1712. https://doi.org/10.1177/1362361320919286

8. Making improvements to Spectrum 10K

The remit of this consultation is to engage with the autism community to find out how the Spectrum 10K study could be improved to make it feel safer and more acceptable. To that end, we deliberately encouraged responses that are focused on improvements rather than on stopping the study. This is a qualitative consultation that looks for the range of feelings and experiences, rather than counting the number of people who believe a certain thing. Thus, the results cannot be used to infer what ‘autism community’ (even if that could be neatly defined) want.

Genetic research is controversial, and the views expressed about Spectrum 10K reflect the highly diverse and conflicted views from autistic people and their families. Controversial topics do not necessarily need to be avoided; there is a place for research that serves the interests of underrepresented subgroups who may be overlooked by majority approaches. Researchers should seek to understand the fears and objections and to consider methods of approach, communication, or engagement that would improve the acceptability of their research. The recommendations in this report are intended to support that endeavour but should not be considered exhaustive. Even with the most thorough and sensitive approach, some people will not support any biomedical research for autism.

We recommend that researchers at the Autism Research Centre at Cambridge and at other institutions do not shy away from such dilemmas, but consider them openly and with full engagement with the communities they research.

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Links to online resources

The following are the links included in footnotes in the text.

https://www.autismresearchcentre.com/

https://wellcome.org/

http://autisticinertia.com/

http://www.autscape.org/

http://www.a2i.co.uk

https://aucademy.co.uk/2021/09/03/boycott-spectrum-10k-please-sign/

https://gizmonaut.net/blog/autism/2022/11/spectrum10k-grant-application.html

Acknowledgements

Appendices

Appendix 1: Full list of recommendations

Autism community and relationship with researchers

- Consider the recommendations for communication detailed in Section 7. Specifically, provide clear, explicit, consistent and detailed information about:
    - The research team, their role in Spectrum 10K, and a statement about their goals and values regarding genetic research
    - The source of funding for Spectrum 10K and the likelihood of commercial application of the results,
    - The motivations of funders and researchers.
- Apologise publicly for insensitive comments.
- Respond quickly to questions from the public about the study. Delays can raise suspicions that there is something to hide.
- Consider the recommendations for involvement detailed in Section 7, in particular:
    - Consider recruiting one or more autistic researchers and/ or expert consultants.
    - Include a wide variety of autistic people and other stakeholders (e.g. family members).
    - Seek out those who bring multiple perspectives, e.g. autistic people who are also parents of autistic children, representatives of autistic people's organisations, and those who have multiple marginalised identities (e.g. ethnic minorities).

Genetic contributions to autism

- Spectrum 10K should communicate its aims openly, ensuring these are clear and consistent. Their meaning shouldn't be lost if they are summarised or adapted for different audiences.
- It is important to recognise that concern about Spectrum 10K's role in the development of a prenatal test for autism hasn't yet been fully addressed. Publicly commit to the stance against prenatal testing for autism.
- Maintain a clear stance against prenatal testing for autism and any research that points in this direction. Amplify the voices of autistic people concerned by developments around this.
- Be a positive public advocate for autism – actively communicate to increase societal awareness that autism doesn't need to be cured or prevented.
- When assessing, documenting and reporting risks, include the risks for the population being studied, i.e., for autistic people more broadly and not just for the individual participants.
- Be clear in publicity and participant information about how GWAS data that will be published can contribute to polygenic risk scores and how these may be (mis)used.

- Ensure that there is a clear ethics statement as part of any publication discouraging the use for prenatal testing or selective termination, and emphasising how little these genes contribute to autism overall.
- Make it possible for participants to consent to each future use of their data.
- Convene a DAC to review requests from other researchers for access to Spectrum 10K data. Specific recommendations for the composition, remit and policies of this committee are detailed in Section 3.5.

Further aims and potential benefits of the study

- Avoid unhelpful labels. Recognise the history of labelling and grouping autistic people and the harms this has caused in the past.
- Clarify what is meant by subgroups, how they will be determined, and how they differ from previous approaches to subgrouping within the autistic spectrum.
- Make it possible for study participants to opt-in separately for subgrouping research.
- Continue to consult autistic people about the design and implications of subgrouping.
- Words such as 'treatment' should be used carefully and explicitly in connection with the aspects of autistic people's lives that they are interested in receiving treatment for (e.g., specific co-occurring conditions).
- Be clear about what benefit may come from subgroups. Ensure that such descriptions are clear, balanced and provided before consent is obtained. Specific information that would be valuable includes:
    - The path from this basic research with no direct benefit to possible future benefits.
    - The intended practical impact of any genetic discoveries about autism (e.g., treating co-occurring conditions, diagnosis).
    - What benefits are not likely, even when that is to the detriment of promoting the study. E.g., would it be expected to lead to social change?
- A complete list of the co-occurring conditions the study intends to investigate should be published and easily accessible.

Data collection and security

- Study questionnaires should be designed in partnership with autistic people to take account of:
    - Including 'it depends' or free text responses where appropriate.
    - Questions that could trigger distress.
    - Stating clearly why the questions are being asked and what benefits the answers will bring.
    - Making the questionnaires accessible to as wide a range of autistic people as possible.
- Make as many questionnaires as possible optional.
- Explore alternative channels to supplying questionnaire information if questionnaires are not accessible/appropriate for some people.
- Explain how discrepancies between questionnaires and medical records are handled.

- Allow participants to view the data held on them in the Spectrum 10K study and add to / amend it.
- Explain clearly which parts of medical records will and will not be accessed and why, make accessing medical records optional, and, if possible, allow participants to specify which parts of their record may be accessed.
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- Be clear about what data is collected, how it is organised, how it is stored, how it will be accessed, how long it will be kept, and what will happen afterwards.
- Create a dynamic Q&A Document in partnership with autistic people that provides clear and unambiguous responses to questions about the methodology of the study and data security, storage and privacy. Updated as questions surface.

Consent and withdrawal

- Report the outcome of discussions with the HRA regarding the inclusion of adults without capacity to consent.
- Require assent from children over age 6 or with the ability to understand the adapted study information.
- Provide information and recording of assent in different formats for accessibility.
- Withdrawal process: consider a two option withdrawal process: No further contact and no further use.
- Explain why data cannot be removed once analysed.
- Recommendations around consent for data sharing have been made in Chapter 3 and for access to medical records in Chapter 5.

Ongoing communication and engagement

- A number of questions posed in the consultation have been set out in this report, and these should be answered in documentation about the study.
- Communicate frequently and openly about Spectrum 10K's developments and about how the study's results have been used.
- Choose language with care: words like 'risk' and 'causes' can be taken as negative and oriented towards prevention.
- Provide clear, explicit, consistent and detailed information.
- Arrange for review of communications by autistic people with relevant expertise prior to publication.
- Produce all study documentation in multiple formats to facilitate access.
- Create opportunities for ongoing involvement and consultation on this and other projects, and advertise these widely.
- Seek to include the widest possible range of views and experiences, including those who are critical of genetic research and those who are negative about being autistic.