



# Spectrum 10K Themes and Responses to Questions

This is the full response by Spectrum 10K on comments and questions raised about the study. Comments and questions were raised about Spectrum 10K:

- When the study was launched
- During [Phase 1 and Phase 2](#) of the consultation

Spectrum 10K's **full response** to these comments and questions is below.

## Contents

Spectrum 10K Themes and Responses to Questions .....	1
A note on ethics approval.....	4
A. Study aims and design.....	4
1. What exactly are the study aims? .....	4
2. Why aren't the aims on the website exactly the same as stated in the grant? .....	5
3. Why does Spectrum 10K need DNA? .....	5
4. Why are you exploring genetic factors and why is it important?.....	5
5. What are the aspects of autism that you will be investigating using Spectrum 10K data? .....	6
6. Why isn't Spectrum 10K collecting DNA from non-autistic people? .....	6
7. Are there any commercial aims and/or potential for diagnostic tests? .....	7
8. Why does Spectrum 10K need questionnaires (including those addressing sensitive issues)? .....	7
9. How will wider societal and environmental factors, including race, gender disparities and health inequalities, be taken into account during the study? .....	8
10. Will the study seek to better understand lived experience of autism? .....	8
B. The value of the study.....	8
11. Is this study going to benefit autistic people or improve wellbeing? If so, how? .....	8
12. How will the study recognise and celebrate positive aspects of autism? .....	9
13. Why isn't the funding being used for things like support and services? .....	9
14. Will the study have any wider benefits to support or education? (Balance between medical and social outcomes?) .....	9
C. The values and the ethical positions of the researchers .....	10
15. What are the researchers' motives for conducting the research?.....	10
16. Do the researchers want to cure or prevent autism? .....	10



17.	Do previous writings/comments contradict stated Spectrum 10K values and aims?.....	11
D.	Funding.....	12
18.	Who is funding Spectrum 10K? .....	12
19.	Is there any funding from the United States? .....	13
20.	What are the non-negotiable aspects of the study? .....	13
21.	What obligations does Spectrum 10K have to its funders? .....	13
22.	Who will benefit financially from Spectrum 10K?.....	13
E.	Who is involved?.....	13
23.	Who is involved in Spectrum 10K and in what capacity?.....	13
24.	Is Autism Speaks involved in Spectrum 10K? .....	14
25.	What was Daniel Geschwind's role in Cure Autism Now (which later became Autism Speaks)? .....	15
26.	Has the ARC ever received funding from Cure Autism Now? .....	15
F.	Genetic Research and concerns about eugenics.....	16
27.	What exactly will Spectrum 10K do with genetic data?.....	16
28.	How does this study build on or relate to other studies into the genetics of autism?.....	16
29.	Even if Spectrum 10K is not looking for a prenatal test, could its findings lead to a prenatal test in the future?.....	17
30.	Can you guarantee that our data will be safe and never get into the hands of those who would wish to harm us?.....	17
31.	Is a prenatal test for autism possible? .....	18
32.	Can autism be diagnosed using genetics alone? .....	18
33.	What do we already know about the genetics of autism?.....	18
34.	If Spectrum 10K does not support the development of a prenatal test for autism, why does the Autism Research Centre support early identification and screening for autism, postnatally?.....	19
35.	Will sub-grouping lead to segregation/discrimination?.....	19
36.	How do you believe that genetic data will be used to help autistic people? How does understanding genetic factors help you to support autistic people and their families? .....	20
G.	Data Use/ Data Sharing/Data Access .....	20
37.	Will Spectrum 10K sell data?.....	20
38.	What data will be shared and why? .....	21
39.	How exactly will decisions about who can access Spectrum 10K data be made? .....	22
40.	Will autistic people have a say on who gets access to the data? .....	23



41.	How <i>exactly</i> will Spectrum 10K data be stored and how secure will this be?	23
42.	Will participants have access to their own data? .....	23
43.	How is the information I give you kept confidential and anonymous? ...	24
44.	What happens to samples and data if somebody withdraws from the study?	24
45.	What will happen to Spectrum 10K data and samples after the project is finished? .....	25
46.	What does it mean that Spectrum 10K is a re-contactable resource for all types of research (genetic or otherwise)? .....	25
H.	Medical records .....	26
47.	Why do you need access to my medical health records (also called electronic health records or NHS health records)? .....	26
48.	How will you access my medical records? .....	27
49.	What will my data be used for? .....	29
50.	How will the study account for possible inaccuracy in medical records?	29
51.	Is sharing medical records compulsory? .....	29
I.	Additional ethical questions based on inclusion of children .....	30
52.	What is the consent procedure for including children in Spectrum 10K (including withdrawal and re-consent)? .....	30
53.	What is meant by 'assent'? .....	30
54.	What will happen to DNA once a child becomes an adult? .....	31
55.	Is it ethical to use DNA from children based on consent from their parents/carers, given that this cannot be retracted if the child decides later that they do not consent? .....	31
J.	Inclusion of adults without capacity to consent to participate in the study.	32
56.	Why is it important to include adults without capacity to consent to participate? .....	32
57.	How does Spectrum 10K include adults without capacity to consent to participate? .....	32
58.	How has participation of autistic individuals with limited communication to participate been designed? How has this been vetted? .....	32
59.	How can capacity to consent to participate in Spectrum 10K be assessed? .....	32
60.	Is it ethical to use DNA from adults who lack the capacity to consent to participate, given that this cannot be retracted if the person is ever able to review the decision for themselves? .....	32
K.	Ambassadors .....	33
61.	What is the ongoing role of Ambassadors in Spectrum 10K? .....	33



L. Autism community involvement prior to pause .....	33
62. How were autistic people involved in the design and setup of Spectrum 10K? 33	
63. Why were relatives and other stakeholders involved? .....	34
64. What are you going to do to engage the autism community more widely? 34	
65. What issues will be covered when you engage the autism community more widely? .....	35
M. Next Steps .....	35
66. When will the study restart? And how long will it take to complete? .....	35

Before we begin:

## *A note on ethics approval*

The Spectrum 10K study was approved by a Research Ethics Committee in England ([London Queens Square REC](#)), and a second committee in Scotland ([Scotland A-REC](#)). Both ethics committees specialise in reviewing research involving vulnerable groups, such as adults who lack capacity to consent on their own.

In addition, approval from the [Health Research Authority](#) (HRA) was also obtained. As standard procedure, the HRA is reviewing the concerns raised with them about the study. Spectrum 10K is in communication with the HRA and the two RECs to provide further information as we progress through the consultation, but the HRA and ethics committees recognise that the original ethical approval still stands.

## *A. Study aims and design*

### *1. What exactly are the study aims?*

#### **The aims of Spectrum 10K**

Spectrum 10K aims to recruit 10,000 people with a formal diagnosis of autism and, where possible, their relatives, to identify genetic and environmental factors that contribute to autism and the physical and mental health of autistic people.

Spectrum 10K will add to basic understanding of the causes of autism and of the link between autism and co-occurring conditions. Beyond this study, the results may have implications for treatment of co-occurring conditions, and identifying who may benefit from different kinds of support. Spectrum 10K also aims to explore the impact that both genetic and non-genetic (environmental) factors have on autism.



## The general aim of our autism research

As researchers, we want to better understand autism, including factors that influence both the positive aspects and the challenges of autism. We hope that by providing evidence-based information about autism, this will lead to improved understanding of autism, including the predictors of wellbeing of autistic people. This could also lead to the development of improved interventions for autistic people who want them.

## Differences and tailored approaches

We know that no two autistic people are exactly alike; some want help in areas such as sensory sensitivity, social interaction, or anxiety, for example, while others do not. In the longer term, understanding these differences, and the biology that underpins them, could help the development of tailored support or treatments for those who want them. For example, future research could use these findings to develop treatments for autistic people who want help with co-occurring conditions like epilepsy, gastrointestinal pain, sleep disorders and anxiety.

### *2. Why aren't the aims on the website exactly the same as stated in the grant?*

Our grant application was written long before we created our website, and some aspects of our thinking about the study evolved after early documents were written, such as the decision to include measures of wellbeing, following consultation with our existing Advisory Panel of autistic people, carers and autism professionals.

In addition, our website was written with a public audience in mind, and therefore uses different language to that found in grant applications.

### *3. Why does Spectrum 10K need DNA?*

Spectrum 10K will explore genetic and environmental factors that contribute to autism and to the physical and mental health of autistic people. Exploring genetic factors requires DNA, which is collected, in this case, via a saliva sample.

### *4. Why are you exploring genetic factors and why is it important?*

We know that autism is partly genetic, but genetically very complex. Many co-occurring conditions such as epilepsy and severe gut problems are also partly genetic and are much more common in autistic people than in non-autistic people. There is a genetic overlap between autism and many co-occurring conditions. We



want to understand the genetic relationship between autism, and the co-occurring conditions.

The causes of these co-occurring conditions may also be different in autistic than in non-autistic people. This also means that autistic people may respond differently to non-autistic people to treatment for these conditions. Therefore, understanding the genes underlying these conditions in autism can help develop better medical support targeting these medical conditions in autistic people.

Finally, there is currently little information available about what works for each person, which means that autistic people may spend years trying different options. Identifying genetic and environmental factors may in the long-term help to better predict which treatments would be best for any given autistic person.

### *5. What are the aspects of autism that you will be investigating using Spectrum 10K data?*

As a part of Spectrum 10K we want to identify genetic variants that are more or less frequently seen in autistic people and investigate how these are linked to co-occurring health conditions. Examples of some questions that can be investigated using the Spectrum 10K data are:

- Understanding whether specific sets of genes contribute to epilepsy in autistic people.
- Understanding whether different genes associated with epilepsy contribute to different types of epilepsy.
- Identifying genes that may contribute to severe gut issues in autistic people. One example is the gene CHD8. Autistic people with this gene are more likely to have severe gut difficulties than those without it, primarily because the nerves in the gut may work differently. Others may have different reasons for severe gut issues. Understanding the genes underlying medical conditions like this may later help scientists develop better medical support for these medical conditions in autistic people.
- Investigating how early life vulnerability interacts with genetics to contribute to depression, anxiety, and wellbeing in autistic people.

### *6. Why isn't Spectrum 10K collecting DNA from non-autistic people?*

We are collecting DNA (and questionnaire data) from non-autistic relatives of autistic Spectrum 10K participants. This is important because it will help us to identify genetic variants that are unique to an individual but not shared with other family members. By comparing differences in DNA with differences in responses to questionnaires, we may be able to explain why one person has a co-occurring condition whilst others do not.





We do not need to collect DNA from non-autistic people because there are already large datasets containing this information, such as the UK Biobank. Many of the participants who contributed to these datasets provided consent to use the data for a broad range of studies, such as Spectrum 10K.

### *7. Are there any commercial aims and/or potential for diagnostic tests?*

Spectrum 10K does not have any commercial aims, and nor does it aim to develop a diagnostic test for autism.

### *8. Why does Spectrum 10K need questionnaires (including those addressing sensitive issues)?*

Questionnaires are helpful because they can help us obtain information about a person's/child's life directly from them or their carer. The questionnaires used in Spectrum 10K cover many aspects that impact the lives of autistic people.

The registration questionnaire for Spectrum 10K includes questions about mental health, physical health, lifestyle, and autistic traits. We also ask these questions about non-autistic relatives so that we can compare information obtained from autistic people with their non-autistic relatives. This will help us to understand what is unique to autistic people and what is shared among family members.

Optional questionnaires have been carefully selected to ask questions about autism that we think will be valuable to autistic people. For example, one questionnaire covers the under-studied topic of sensory sensitivity differences.

Some of the optional questionnaires include sensitive questions related to mental and physical health. While some include potentially distressing topics such as experience of child abuse or feelings of suicide, which participants may find upsetting, we believe it is important to study these topics. Asking questions about suicide may lead to ways of helping people in the future. We will always warn participants about potentially distressing questions and provide information about where to seek help if they do become distressed. These are optional questionnaires and participants have the option to opt out completely or skip individual questions they are not comfortable answering.

The more information we analyse, the better we will be able to understand the diversity of experiences within autism and co-occurring physical and mental health conditions.



*9. How will wider societal and environmental factors, including race, gender disparities and health inequalities, be taken into account during the study?*

Whilst we cannot capture all of wider societal and environmental factors, we have endeavoured to capture some of these important factors using self-report questionnaires. For example, we ask people about any stigma and discrimination they have faced in one optional questionnaire. We also collect information about gender, ethnicity, educational attainment and income, to incorporate these into our analyses.

*10. Will the study seek to better understand lived experience of autism?*

Spectrum 10K is a quantitative research study because of its scale, and the questionnaires we use aim to capture autistic people's lived experience across a range of areas, as much as is possible within the limits of this type of research.

*B. The value of the study*

*11. Is this study going to benefit autistic people or improve wellbeing? If so, how?*

There are no direct benefits of taking part. However, Spectrum 10K will help to further our understanding of the relationship between genes, environment, autism and related conditions.

Spectrum 10K will help scientists better understand how genetic and environmental factors affect the wellbeing of autistic individuals, including their physical and mental health. In the longer term, after Spectrum 10K has ended, we hope this increased understanding will lead to improvements in the quality of support and care for autistic people and their families. This will require further research studies after Spectrum 10K has completed, and that work will not necessarily be completed by the researchers involved in Spectrum 10K.

The knowledge gained from Spectrum 10K will help promote greater understanding into the causes of the wide range of outcomes in autistic people including their strengths and talents. Findings from Spectrum 10K may inform future studies to identify who might benefit from different kinds of interventions, if they are seeking them.



## *12. How will the study recognise and celebrate positive aspects of autism?*

Spectrum 10K aims to collect data about mental and physical health outcomes. Working within the neurodiversity paradigm, we emphasise that autism is characterised by differences, including strengths and talents. Examples that we study include measures of excellent attention to detail and memory for detail, and excellent pattern recognition.

We do also recognise that many autistic people understand autism to be a disability, so we also include questions about language development, social understanding, and learning disability in our questionnaires.

The language we use emphasises differences in both the mind and biology, rather than now outdated language referring to disorders or pathology, that could stigmatise. We hope that the language we use both when communicating with participants and when sharing research with scientific audiences will avoid stigmatising views of autism and instead celebrate the positive aspects of autism as one example of neurodiversity.

## *13. Why isn't the funding being used for things like support and services?*

This study is funded by the Wellcome Trust. The Wellcome Trust is a charitable organisation that funds research into human health.

We support research into services and support for autistic people. While the Wellcome Trust funding cannot be used for these purposes, other bodies provide funding for this kind of research. We hope that funding for support and services will be increased in the future as we recognise that support services are inadequate and under-funded. This is true in both the NHS and Social Services. We are playing our part in trying to bring in more funding for support services through our charity, the Autism Centre of Excellence (ACE) at Cambridge.

## *14. Will the study have any wider benefits to support or education? (Balance between medical and social outcomes?)*

Spectrum 10K is focused on health outcomes. It is unlikely that Spectrum 10K will directly benefit social support or education.



## *C. The values and the ethical positions of the researchers*

### *15. What are the researchers' motives for conducting the research?*

The researchers aim to understand the genetic and social factors contributing to autism and co-occurring conditions. See below for our values and ethical positions.

### *16. Do the researchers want to cure or prevent autism?*

No. We view autism as a form of neurodiversity. We believe that all forms of neurodiversity should be celebrated. We do not support cures or treatments for autism itself.

We do recognise that some autistic people are distressed by some aspects of autism, such as hypersensitivity, and some conditions that commonly co-occur with autism, such as epilepsy or chronic anxiety. We would consider researching these areas (but not autism itself) for possible interventions or even cures.

We also recognise that some of the conditions that co-occur with autism are severe neurodevelopmental conditions like DDX3X Syndrome, Wiedemann-Steiner Syndrome, and Kabuki Syndrome. Severe neurodevelopmental conditions may cause developmental delay, intellectual disability, motor coordination difficulties, or even impaired organ development. Sometimes, a lack of clarity in terminology means that autistic people who have a co-occurring severe neurodevelopmental condition are only referred to as "autistic" without mentioning their additional severe neurodevelopmental condition. This means that research into treatments for severe neurodevelopmental conditions is sometimes misunderstood as research into treatments for autism. We recognise that treatments and improved clinical management for some severe neurodevelopmental conditions may be needed for some symptoms in some individuals. However, we distinguish this from seeking cures and treatments for autism, which we do not support.

We also state categorically that we do not support the prevention of autism.

We understand that, as part of clinical care, the NHS offers prenatal testing to some parents who have already had a child with a severe neurodevelopmental condition and may wish not to have a second child with the same condition. We do not wish to comment on parental choices to prevent severe neurodevelopmental conditions but reiterate that we distinguish such conditions from autism.



## 17. Do previous writings/comments contradict stated Spectrum 10K values and aims?

No members of the research team have published opinions or are conducting research that contradicts the aims: "Spectrum 10K aims to investigate genetic and environmental factors that contribute to the wellbeing of autistic individuals and their families."

We are aware that some members of the public believe our researchers are seeking a prenatal test for autism as a way to prevent autistic babies from being born in the future. We have categorically stated that this is not the case. Aspects of some published articles appear to have been misinterpreted or taken out of context.

One example relates to an article published by Professor Simon Baron-Cohen in 2009, which includes the line "Caution is needed before scientists embrace prenatal testing so that we do not inadvertently repeat the history of eugenics or inadvertently 'cure' not just autism but the associated talents that are not in need of treatment." This article has been interpreted by some as meaning that Professor Baron-Cohen was, at that time, in support of a prenatal test and cure for autism and concerned only about the preservation of 'associated talents'. In fact, the opposite is true.

Professor Baron-Cohen clarifies that he has never been in support of a cure for autism and has never been in favour of a prenatal screening test for autism. Indeed, his article in 2001 was one of the first to question why autism was seen as a disorder, disease or even a disability, and argued strongly for recognizing autism as a difference. He was the first person to introduce the term Autism Spectrum Conditions (ASC) instead of Autism Spectrum Disorder (ASD), to shift perception away from disease or disorder and towards difference and disability. His intention in the 2009 article mentioned above was to encourage scientists to question the then widespread assumption that autism needed a cure. He was at the forefront of encouraging scientists and clinicians to question this.

Professor Baron-Cohen has explained that in highlighting how 'cures for autism' could also lead to a loss of talents, he was making the point that autism entails both strengths as well as challenges. In another 2009 article, in *Community Care* magazine, he clearly stated his own position that all autistic people's lives have value, whether or not they have talents. In response to a *Guardian* article misrepresenting ARC research as developing a prenatal test for autism (the ARC has never conducted research with the aim of developing a prenatal test for autism) Professor Baron-Cohen wrote a series of commentaries in *New Scientist* to correct the record. More recently, he has reiterated both his and the ARC's opposition to eugenics and to viewing autism as a disease or disorder in need of a cure. His keynote speech at the UN in New York on Autism Awareness Day 2017 was one of the first to address the topic of autism and human rights, and his



record over many decades shows his commitment to respecting autistic people. He is outspoken in his support of neurodiversity.

It is also important to note that the Spectrum 10K Principal Investigators make a distinction between autism and severe neurodevelopmental conditions, the latter being conditions in which the individual may have developmental delays, intellectual disability, epilepsy, and/or motor coordination difficulties, as well as potentially impaired development of other organs, e.g. congenital heart defects. Examples are DDX3X syndrome, Wiedemann-Steiner Syndrome (KMT2A), and Kabuki syndrome (KMT2D). Cures, treatments and improved clinical management for some conditions in this category may be needed for some symptoms in some individuals. The PIs are aware that some parents who already have one child with such conditions may request prenatal testing because they wish to avoid having a second child with the same condition. The PIs see this as outside of their roles as scientists and are aware that this use of prenatal testing is provided as part of clinical care within the NHS

(<https://www.genomicseducation.hee.nhs.uk/blog/what-is-nipd/>,  
<https://bwc.nhs.uk/prenatal-reproductive-genomic-testing/>).

It would be wrong for the PIs to comment on parental choice to terminate a pregnancy, but the PIs encourage more discussion and debate in the public on these bioethical issues. To reiterate, it is important to distinguish seeking a cure or treatment for such co-occurring conditions, which may be wanted as part of medical care, and which the PIs do support, vs. seeking a cure or treatment for autism itself, which the PIs do not support. Finally, and again to be clear, none of the PIs support prevention of autism.

There is also a lack of clarity in terminology, perhaps more in the US than in the UK. At times, those with severe developmental conditions and autism are referred to only as 'autistic', rather than as having severe developmental disorders *and* autism. This can lead to misunderstandings, and research seeking a cure or treatment for severe neurodevelopmental conditions can be misconstrued as seeking to cure autism. We hope we have been clear that none of the PIs are seeking to prevent or cure autism.

## D. Funding

### 18. Who is funding Spectrum 10K?

Spectrum 10K is wholly funded by the Wellcome Trust. The Wellcome Trust is an organisation that provides funding to scientists to support them with research that can increase understanding of physical and mental health and ways to improve them. For more information about the Wellcome Trust, visit [www.wellcome.ac.uk/about-us](http://www.wellcome.ac.uk/about-us).



## *19. Is there any funding from the United States?*

Spectrum 10K is not receiving any funding directly or indirectly from the United States or any other countries. All of the funding is provided by the Wellcome Trust, which is based in the United Kingdom. The Wellcome Trust generates its funding through its own financial investments around the world and generally does not receive money from other organisations. For more information about where the Wellcome Trust's money comes from, visit [wellcome.org/who-we-are/investments](http://wellcome.org/who-we-are/investments).

## *20. What are the non-negotiable aspects of the study?*

Please refer to the limits of the consultation [document](#).

## *21. What obligations does Spectrum 10K have to its funders?*

The Spectrum 10K research team has obtained the funding from the Wellcome Trust. Our obligations to our funders are to establish a cohort of 10,000 autistic people, and where possible their families. We will also use this cohort to investigate genetic and environmental links to autism and co-occurring conditions.

## *22. Who will benefit financially from Spectrum 10K?*

None of the researchers, members of the research team, the funders or anyone else stand to benefit financially from Spectrum 10K.

## *E. Who is involved?*

## *23. Who is involved in Spectrum 10K and in what capacity?*

The Spectrum 10K research is being led by a team from the Autism Research Centre (ARC), which is part of the University of Cambridge.

The Principal Investigator (PI) for Spectrum 10K is Professor Simon Baron-Cohen, who is the Director of the ARC. As the PI Simon oversees and is responsible for all aspects of Spectrum 10K, including overseeing and directing the studies design and scientific integrity.

The team also includes four Co-Principal Investigators (Co-PIs) from the Wellcome Sanger Institute, which is a research institute in the United Kingdom, and from the University of California Los Angeles (UCLA), which is a public university in the United States. Professor Simon Baron-Cohen and the Co-PIs share responsibility for data analysis and the scientific integrity of the project.



The Co-Principal Investigators are:

- Professor Daniel Geschwind, who is the Gordon and Virginia MacDonald Distinguished Professor of Human Genetics, Neurology and Psychiatry at UCLA.
- Professor Matthew Hurles, who is the Head of Human Genetics at the Wellcome Sanger Institute.
- Professor Hillary Martin, who is a Group Leader at the Wellcome Sanger Institute.
- Professor David Rowitch, who is the Head of Paediatrics at the University of Cambridge.

The Spectrum 10K team also involves the Director of Research Strategy, study co-ordinators, a post-doctoral researcher, and a data manager – all based at the ARC.

For more information about the research team, visit [spectrum10k.org/our-team/](http://spectrum10k.org/our-team/).

Spectrum 10K is sponsored by the University of Cambridge and by the Cambridgeshire and Peterborough NHS Foundation Trust (CPFT). The sponsors take overall, legal responsibility for ensuring that Spectrum 10K is set up, runs, and reports appropriately.

Spectrum 10K is also working with 18 NHS Trusts across the UK. The role of these Trusts is to help identify people who would like to participate in Spectrum 10K and to help them register for the study.

### **Consultation Team**

The Spectrum 10K consultation is being co-led by Hopkins Van Mil, which is an independent agency, by Leneh Buckle, who is an autistic researcher working on a freelance basis for Hopkins Van Mil, and by the Spectrum 10K team based at the ARC.

## *24. Is Autism Speaks involved in Spectrum 10K?*

Autism Speaks is not involved in Spectrum 10K. Autism Speaks is not providing any of the funding for the project. We have not discussed the project with Autism Speaks at any point.

During Spectrum 10K, we plan to analyse previously collected genetic data from several other databases. One of the databases that we plan to use is the Autism Genetic Resource Exchange (AGRE), which is owned by Autism Speaks. We will have to apply to Autism Speaks for approval to access AGRE and pay a small fee to Autism Speaks for each piece of data that we access. This will not enable Autism Speaks to influence Spectrum 10K in any way other than approving or denying our application. For more information about the process for accessing



AGRE, visit <https://www.autismspeaks.org/applying-access-agre-data-and-biomaterials>.

Some members of the research team have an indirect relationship with Autism Speaks. The ARC and the Spectrum 10K PI, Simon Baron-Cohen, are involved in AIMS-2-TRIALS, which is a research project being carried out by 48 European organisations. Whilst AIMS-2-TRIALS is primarily funded by the European Union, it receives some of its funding from Autism Speaks. Spectrum 10K is not directly involved with AIMS-2-TRIALS in any way. For more information about Autism Speaks' role in AIMS-2-TRIALS, visit

<https://www.aims-2-trials.eu/about-aims-2-trials/team/autism-speaks/>. In addition, in 2010 one graduate student at the ARC was funded by a predoctoral fellowship investigating 'Hypersensitivity in autism: a psychophysiological and neuroscientific approach', which was funded by Autism Speaks. The fellowship ended in 2012 and the ARC has received no direct funding from Autism Speaks since then.

## *25. What was Daniel Geschwind's role in Cure Autism Now (which later became Autism Speaks)?*

In the 1990s Professor Daniel Geschwind chaired Cure Autism Now's (CAN) scientific advisory board as a neurologist.

Cure Autism Now was created by parents in the autism community in the 1990s to fund autism research, primarily for syndromic forms of autism (autism with a known genetic cause, such as Fragile X syndrome and Tuberous Sclerosis Complex), at a time when there was almost no funding.

Daniel Geschwind's research was never funded by CAN. He did not receive money from CAN for his involvement.

Daniel Geschwind shares the values of the rest of the Spectrum 10K team. He acknowledges that language, concepts and priorities of the autism community have changed over the past 30 years.

## *26. Has the ARC ever received funding from Cure Autism Now?*

The Autism Research Centre received a grant for approximately £50,000 from Cure Autism Now (CAN) in 2002. An example of research funded by this grant is this functional MRI study looking at cognition in autistic people. A further grant for approximately £62,000 funded by Cure Autism Now, 'The role of perceptual attentional disturbances in the early diagnosis of autism', ended in 2008. At the time of receiving CAN funding, CAN was one of the few funding bodies available in the UK.



## *F. Genetic Research and concerns about eugenics*

### *27. What exactly will Spectrum 10K do with genetic data?*

All the saliva samples that we receive will be provided with an ID number and stored separately from participants' personal details, so that no participant can be identified from their samples or genetic data. This is called pseudonymisation. From the pseudonymised saliva sample, a laboratory will extract DNA.

For each sample, Spectrum 10K team members will analyse some or all of the DNA code along with the participant's anonymised clinical data. We will be searching for structural and functional changes in the DNA that may contribute to autism and related conditions.

Spectrum 10K will only use genetic data for the aims of the study: to identify genetic and environmental factors that contribute to autism and to the physical and mental health of autistic people. We will not be using this genetic data to develop a genetic test for autism.

We will also store the DNA samples for 10 years and genetic data obtained from the samples for 25 years. In the future, we may share the data with trusted academic or commercial collaborators who are conducting ethically approved research in line with the aims and values of Spectrum 10K.

### *28. How does this study build on or relate to other studies into the genetics of autism?*

There are a few ongoing and previous studies of autism that have collected and analysed DNA from autistic people. Some of them have also collected and analysed DNA from parents of autistic children. Examples of such studies include SPARK and the Simons Simplex Collection. These studies have demonstrated that autism is genetically very complex, with 1000s of genes involved in different combinations which contribute to autism and co-occurring conditions.

Spectrum 10K builds on these existing studies. Spectrum 10K is different from almost all previous autism genetic studies in two ways. First, it will collect richer questionnaire data from participants including questions related to their health conditions, their education, stigma and vulnerabilities, quality of life, and detailed questions on some mental health conditions. Second, Spectrum 10K aims to obtain data from electronic medical records. These additions help us to better understand how genetic and environmental factors are linked to co-occurring conditions in autism.



## *29. Even if Spectrum 10K is not looking for a prenatal test, could its findings lead to a prenatal test in the future?*

It is highly unlikely that genetics alone could ever be used to develop a prenatal test for autism. Thousands of genetic changes contribute to the likelihood of someone being autistic and it is impossible to diagnose someone as autistic based only on their genes. For example, there are identical twins who share 100% of their DNA but where only one twin is autistic. This demonstrates that there are non-genetic (e.g., environmental) factors that contribute to the development of autism.

Autism cannot be diagnosed using only genetics. Autism is only partly genetic and there is no single gene for autism. Autism is genetically complex.

Finally, autism is diagnosed on the basis of difficulties in social communication and in adjusting to unexpected change, and these can only be observed in behaviour postnatally.

## *30. Can you guarantee that our data will be safe and never get into the hands of those who would wish to harm us?*

Your data will be kept strictly confidential. Any data you share with us will be stored under a unique ID in a process known as pseudonymisation. This means that you cannot be identified from any of the samples or data you provide.

Spectrum 10K data are being securely stored on a University of Cambridge safe-haven (ISO27001 accredited). Your data will be stored on secure servers located at the University of Cambridge and will only be accessible by approved Spectrum 10K researchers.

We will co-design a sharing policy with the autism community so that, eventually, data from Spectrum 10K participants can be shared with collaborators in the spirit of advancing research. It will not be shared with anyone to develop prenatal testing for autism or a cure for autism.

Your anonymous medical and health data will be stored separately from your personal details.

All data collected will be collected in line with the Data Protection Act 2018 and GDPR (General Data Protection Regulations).

Data collection will also comply with the Common Law Duty of confidentiality. You can read more about it at:

<https://researchgovernance.medschl.cam.ac.uk/information-governance-storage-of-research-participant-data/secure-data-hosting-policy/>



We have mechanisms in place to keep your data safe. One of these is that we will set up a Data Access Committee, made up of autistic people, their family members, and scientists, to ensure that any scientist who applies for access to our data has values that align with ours, including opposing the development of a prenatal test for autism, opposing the prevention of autism, and opposing the development of a cure for autism.

### *31. Is a prenatal test for autism possible?*

We do not think autism can be diagnosed prenatally. Autism is complex, and two autistic people may be very different from each other. In some people, signs of autism may only be clear later on in life. There is no single biological marker for autism, and as such, we do not think anything that can be measured prenatally can diagnose someone as being autistic.

### *32. Can autism be diagnosed using genetics alone?*

No. Autism cannot be diagnosed using only genetics.

Autism is only partly genetic and there is no single gene for autism.

Autism is genetically complex. Thousands of genetic changes contribute to the likelihood of someone being autistic and it is almost impossible to diagnose someone as autistic based only on their genes. For example, there are identical twins who share 100% of their DNA but only one twin is autistic.

This contrasts with Down's Syndrome. Down's Syndrome is a genetic condition caused by an extra copy of chromosome 21 and can be easily detected by looking at a person's genes.

### *33. What do we already know about the genetics of autism?*

A lot has been accomplished, but there is still a lot to learn. The genetics of autism is complex.

We know there are millions of genetic variants (a change in the DNA sequence making up a gene), which each increase a person's likelihood of being autistic by a very small amount.

Scientists worldwide have only found about 5 common genetic variants that are associated with autism. This is when a gene variant is found in more than 1% of people. Everyone carries some of these common genetic variants.

Particular combinations of these variants increase the likelihood that a person will be autistic.

However, the common genetic variants we know about only explain about 2% of the likelihood that a person is autistic.



If we found all common genetic variants, this would still only explain about 40% of the likelihood that someone is autistic. Scientists believe that another 10% of the likelihood is accounted for by rare gene variants. These are genetic variants that are only found in fewer than 1 per 1000 people. Therefore, there must be other non-genetic influences that explain the other 50% of the likelihood that someone is autistic.

DNA alone cannot explain the causes of autism and it is still unclear which environmental factors contribute to autism. Studying both genetic and non-genetic (e.g. environmental) factors will give us a clearer picture.

### *34. If Spectrum 10K does not support the development of a prenatal test for autism, why does the Autism Research Centre support early identification and screening for autism, postnatally?*

We want to see the best possible outcomes for every autistic person.

Early identification of autism should lead to better support, early intervention, avoid delays in diagnosis and prevent common mental health problems such as anxiety and depression from occurring.

This should also allow better clinical management of co-occurring conditions such as epilepsy, gastrointestinal pain, sleep disorders and anxiety.

We know co-occurring conditions, such as gastrointestinal problems, epilepsy, sleep disorders, anxiety and depression may compromise someone's comfort, health or happiness and impact their quality of life. Early identification can help towards greater wellbeing.

We do not think it will ever be possible to diagnose autism using genetic information alone. However, it may be possible to use genetics in combination with other diagnostic assessments postnatally to help diagnose autism and provide more precise information about possible co-occurring health conditions. Finally, Spectrum 10K is not developing a genetic diagnostic test for autism.

### *35. Will sub-grouping lead to segregation/discrimination?*

We are aware that this is a concern for some autistic people. We oppose any discrimination and we are aware that there are reasons for and against using subgroups.

We are aware that different autistic people have very different needs. We need to acknowledge that subgroups already exist. For example, some autistic people have language delay and some do not. Some autistic people have learning disability and some do not. Some autistic people have epilepsy and some do not. Some autistic people have gastrointestinal pain and some do not. We do not see



one subgroup as better than another, as all people are of equal value and should have the same rights. Spectrum 10K hopes to deepen our understanding of existing subgroups and even identify new subgroups to better understand the needs of different autistic individuals. In the long run, this could provide more personalised support.

### *36. How do you believe that genetic data will be used to help autistic people? How does understanding genetic factors help you to support autistic people and their families?*

Genetic data can be helpful in a few different ways, especially for understanding co-occurring conditions. First, genetic data can help identify meaningful subgroups of autistic individuals. This may help us understand who needs different types of support. Subgroups include rare syndromic forms of autism with specific co-occurring conditions. One example is the gene CHD8 where autistic individuals are more likely to have severe gut difficulties.

Some co-occurring conditions like epilepsy and certain forms of gut difficulties are strongly genetic. Understanding the genes underlying these conditions may help develop better medical support targeting these conditions in autistic people.

Other co-occurring conditions like sleep issues, epilepsy, hypermobility, Ehlers-Danlos Syndrome (EDS) and depression are partly genetic, and arise from the interaction between our genes and our environment. Here, capturing genetic information together with environmental factors may identify subgroups of autistic people who may have very different reasons for sleep difficulties and depression. For some of these individuals this kind of research may lead to being able to provide better medical support for these conditions.

A third factor is understanding who will respond to different kinds of support. For instance, people may respond differently to different support for co-occurring conditions such as depression and anxiety. Unfortunately, there is little information to predict how people will respond and what works for different individuals. This means that people may spend years trying different options. Using genetics and environmental factors may help us build better models to predict who responds to different interventions.

## *G. Data Use/ Data Sharing/Data Access*

### *37. Will Spectrum 10K sell data?*

No. We will not sell data at any point, during or after the study. We have no commercial motive for conducting this research.

We may share anonymised data with commercial collaborators who are conducting ethically approved research in line with the aims and values of





Spectrum 10K. We do not want to exclude the possibility that future research may benefit autistic people just because it is being conducted by non-academic companies. Commercial collaborators will go through exactly the same process for requesting access to Spectrum 10K data as will academic collaborators. This process will make sure that any commercial collaboration is in line with the ethical values of Spectrum 10K. Examples of possible commercial collaborations include the development of a drug to alleviate a specific type of epilepsy or gut difficulty relevant to some autistic people. Such research is typically conducted in commercial collaborations.

We recognise that some participants may not wish their data to be shared with commercial collaborators, so this will be optional. At the point of signing up to participate in Spectrum 10K, participants will be asked to tick a separate box to indicate that they consent to their data being shared with commercial collaborators. Participants who do not consent to their data being shared with commercial collaborators will still be able to take part fully in Spectrum 10K.

### *38. What data will be shared and why?*

Only approved members of the Spectrum 10K team will be able to access the Spectrum 10K data. They will only be able to access the data that they need and limited pre-approved access to data will be provided. Only specific researchers in the team will have approval to access personally identifiable data which will be limited to the Database Manager and approved individuals for data management purposes.

In the future, we would like to share Spectrum 10K data with other researchers. This is important for lots of reasons. First, it makes science more transparent by enabling results to be replicated. Second, it prevents other researchers from wasting resources by collecting the same data in future. Third, it maximises the possible public benefits from the data by enabling different researchers with different areas of expertise analysing the same data in different ways. For example, as a parallel, the UK Biobank contains questionnaire, genetic, and medical data from 500,000 adults. Researchers who have been given access to this data have been able to identify people who need regular screening for breast cancer and heart disease.

We will only share anonymised Spectrum 10K data with external researchers who are conducting ethically approved research in line with the aims and values of Spectrum 10K. Only researchers who have been approved by the Data Access Committee will be able to access Spectrum 10K data. They will only have access to the data that they need for their research, never to participants' personally identifiable information, and will only be able to use the data for the purposes of their research.

We will not share any data with the police unless we are legally obligated to do so. We will never share data with employers, local authorities, or anybody else who



will not use the data purely for research purposes approved by the Data Access Committee.

### *39. How exactly will decisions about who can access Spectrum 10K data be made?*

We will co-designing our data sharing policy with the autism community. This will include setting up a Data Access Committee made up of researchers, autistic people, and parents and caregivers of autistic people. The Committee's exact membership, structure, methodology, and decision-making processes will be decided during our consultation with the autism community. We will publish all these details before we ask participants to sign up to Spectrum 10K.

No data will be shared without the approval of the Data Access Committee. To apply for access to Spectrum 10K data, external researchers will have to submit a full proposal to the Committee that describes what their research is, who is involved in the research, what they will use Spectrum 10K data for, and what the value of their research will be for science and the autism community. The Data Access Committee will evaluate the application using a checklist that is being co-designed with the autism community. The checklist will be designed to ensure that the proposed research is consistent with the aims and values of Spectrum 10K and has been approved by an independent research ethics body. We will publish the checklist on the Spectrum 10K website along with the rest of our data-sharing policy and summaries of any approved research projects, so that participants and other members of the public can know about all the research that is being carried out with Spectrum 10K data.

The Data Access Committee (also sometimes referred to as the Data Committee Steering Group) will not approve any access to Spectrum 10K data by researchers who are seeking to develop a prenatal test for, or to cure or prevent, autism.

To be approved for access to Spectrum 10K data, external researchers will have to sign a formal and legally binding data transfer agreement with the University of Cambridge that states explicitly what they can and cannot use the data for. They will also have to agree to submit any scientific papers that they write using Spectrum 10K data to the Data Access Committee for scrutiny before they can submit the paper to an academic journal for publication. This will enable the Data Access Committee to make sure that the research carried out with Spectrum 10K data is consistent with the terms on which access to Spectrum 10K data was originally granted.

#### *40. Will autistic people have a say on who gets access to the data?*

Yes. The Data Access Committee will include autistic people. The exact make-up of the Data Access Committee and the data sharing policy that the Committee will follow when it decides who can access the Spectrum 10K data will both be co-designed with members of the autism community during our consultation.

#### *41. How exactly will Spectrum 10K data be stored and how secure will this be?*

Spectrum 10K data will be stored in a Spectrum 10K database on the University of Cambridge's Secure Data Hosting Service, which is a "Data Safe Haven" with ISO 27001 certification. This means that the data will be fully secure. It is password protected, only accessible by approved members of the Spectrum 10K team, only accessible from virtual desktops without access to the internet, and unable to be copied to any other device or server. This data is pseudonymised, so that researchers will not be able to identify participants while they carry out research.

Genotype data will be stored on the Secure Research Computing Platform (SRCP), which is a secure private tenanted cloud platform for the storage and processing of level 3 confidential data and sits within the University of Cambridge. The platform requires 2-factor authentication, is ISO 27001 certified and is backed up.

Saliva samples are stored and analysed at a licensed laboratory and biorepository. Samples are only labelled with a barcode and the laboratory and biorepository do not have access to any personally identifiable information. This is also the case with samples collected prior to the pause of Spectrum 10K.

When the Data Access Committee approves a collaborator's request to access the Spectrum 10K data, the collaborator will only be able to access the data through a highly secure research database called a "repository". The data will remain secure, controlled by the Data Access Committee, and anonymised, so that the collaborator cannot identify participants from their samples or data. The collaborator will have to sign a formal and legally binding data transfer agreement with the University of Cambridge, which will state explicitly what they can and cannot use the data for. They will not be able to copy or share the data further.

#### *42. Will participants have access to their own data?*

Participants will not have access to their own data. This is because under data protection legislation within the UK (UK GDPR and Data Protection Act 2018), specific exemptions are in place relating to data processing for scientific research and statistical purposes. This is to ensure the validity of the research that takes



place. Data collected in Spectrum 10K is pseudonymised and research participants will not be identifiable from research results.

### *43. How is the information I give you kept confidential and anonymous?*

Protecting the privacy of participants is our top priority. We will store personally identifiable information such as names, addresses, email addresses, and dates of birth separately from non-identifiable data such as saliva samples and medical data.

We will give non-identifiable data a unique ID in a process known as pseudonymisation. This means that researchers will not be able to identify individual participants from their samples or data while they carry out their research.

Only specific researchers in the team will have approval to access personally identifiable data. This will be limited to the Database Manager and one or two approved individuals for data management purposes. We will collect and use the minimum amount of personally identifiable data possible.

All the data will be stored securely and will be collected in line with the Data Protection Act 2018, the General Data Protection Regulations (GDPR), and the Common Law Duty of Confidentiality.

### *44. What happens to samples and data if somebody withdraws from the study?*

Participants will be able to withdraw from the study, without giving any reason, by contacting [info@Spectrum10K.org](mailto:info@Spectrum10K.org).

We modelled our withdrawal at three levels:

#### **No Further Contact:**

This means that Spectrum 10K would no longer contact you directly but would have permission to retain and use information and samples you provided previously and to obtain and use further information from your health records. This level of withdrawal allows researchers to study autism with the goal of improving the health of autistic people and their family members.

#### **No Further Access:**

This means that Spectrum 10K would no longer contact you or obtain further information from health records in the future, but still has permission to use the information and samples provided previously.

#### **No Further Use:**



In addition to no longer contacting you or obtaining further information, any information and samples collected previously would no longer be available to researchers. Spectrum 10K would destroy samples (although it may not be possible to trace all distributed sample remnants) and would only hold information for archival audit purposes. Such a withdrawal would prevent information about you contributing to further research, but it would not be possible to remove data from research that had already taken place.

### **Going Forward**

After further discussion, the Spectrum 10K team would like to propose simplifying this process by reducing the withdrawal levels to only one.

Should a participant wish to withdraw under the proposed simplified withdrawal process, they would be withdrawn at the level of “no further use”. All personally identifiable data would be deleted. We would retain non-identifiable data only for auditing purposes, so that we can ensure the validity of data analysis that has already taken place. This is essential for making sure that our research is reliable and accurate.

#### *45. What will happen to Spectrum 10K data and samples after the project is finished?*

After the project is finished, samples will be stored for 10 years, and data will be stored securely for 25 years. During this period, they may be shared with other researchers. After 25 years the data will be deleted.

#### *46. What does it mean that Spectrum 10K is a re-contactable resource for all types of research (genetic or otherwise)?*

By ‘re-contactable resource’, we mean that we may contact participants in Spectrum 10K to invite them to participate in other ethically approved studies as well. We may re-contact participants about any studies seeking to better understand co-occurring conditions and wellbeing in autism, whether or not those studies are primarily about genetics. Participants will be sent a maximum of 4 invitations per year.

Participants do not have to participate in other studies. They can decide when they receive an invitation to another study whether they would like to participate in that study or not.

We are open to discussion with the consultees about making this aspect of the study optional.



## *H. Medical records*

### *47. Why do you need access to my medical health records (also called electronic health records or NHS health records)?*

One of the aims of Spectrum 10K is to identify genetic and environmental factors that impact on the physical and mental health of autistic people. To achieve this aim, we need to collect information about participants' co-occurring health conditions.

There are two ways we can collect information about co-occurring health conditions:

- Through questionnaires
- Through medical health records (Electronic Health Records) held in the NHS.

We will be able to collect some information about co-occurring health conditions through our questionnaires, which participants will complete when they sign up to participate in Spectrum 10K. However, our questionnaires may not cover all co-occurring health conditions, especially rare conditions.

We can also collect information about co-occurring health conditions by accessing information about participants' diagnoses, prescribed medications, hospitalisations, referrals, and test results from their Electronic Health Records, which are held by the NHS. We set this out within the Spectrum 10K grant application to the Wellcome Trust. A combination of information collected from questionnaires and Electronic Health Records can provide more complete information of someone's health than using either one of them alone.

By analysing participants' genetic and questionnaire data alongside this information from their Electronic Health Records, we will be better able to understand the relationship between genes, co-occurring health conditions, and health outcomes.

We are requesting access to participants' Electronic Health Records because many other research studies have shown how useful this information can be. For example, other researchers have recently used Electronic Health Records to identify groups who are more vulnerable to long COVID or more likely to develop dementia. This makes it easier for health and social care services to support these groups. We would like to do the same for groups of autistic people who might be more likely to have or develop co-occurring health conditions.





## 48. How will you access my medical records?

Your medical records (Electronic Health Records) will be accessed through applications to national bodies who are responsible for data governance and access. There are strict data sharing protocols which are overseen and authorised by national bodies in England, Wales, Scotland and Northern Ireland. Particular organisations oversee and are responsible for governance of data access and applications. In some circumstances, though not always separate organisations oversee the data sharing process. For example: **NHS Scotland** Public Benefit and Privacy Panel for Health and Social Care (**HSC-PBPP**) is a governance structure of NHS Scotland (NHSS) who scrutinise and approve an application. Data access is managed and overseen by electronic Data Research and Innovation Service (**eDRIS**), whose team are part of Public Health Scotland.

Once an application has been completed, submitted to the relevant application body and approved, a signed data access agreement is put in place. Data is then shared electronically via secure Safe Havens. Health records are accessed using participants' personal details and/or their NHS number. Only one person in the Spectrum 10K team will be able to link your DNA and questionnaire data to your NHS data. This is the database manager. As soon as the data has been linked in the Spectrum 10K database, the information is pseudonymised.

### **Can researchers see entire health records?**

No. The Spectrum 10K team will only access the relevant sections of participants' health records. Researchers will make a request, for example asking about additional diagnoses or medication, a data manager at the National Digital Health organisations will then find and extract that information and share *only that information* with researchers.

### **Is it unusual for researchers to access health records?**

Accessing health records in this way is not unique to Spectrum 10K. Many research studies and databases have linked to medical health records to provide crucial information on health. For example, a recent study used medical health records to understand who are more vulnerable to long COVID ([Millions of GP records provide vital insights into long COVID \(digitalhealth.net\)](https://www.digitalhealth.net)).

Similarly, using medical health records, researchers have been able to identify those who are more likely to develop dementia ([Electronic health records-based tool uses data to detect undiagnosed dementia | National Institute on Aging \(nih.gov\)](https://www.nih.gov))

For further information on the national organisations responsible for governance and data access in England, Wales, Scotland and Northern Ireland, please see the following:

### **England:**



NHS England is the custodian for health and care data for England. Applications to link health data are submitted to the Data Access Request Service (DARS) within NHS England.

Information about how NHS England protects your data can be found here:

<https://digital.nhs.uk/data-and-information/keeping-data-safe-and-benefitting-the-public/how-we-look-after-your-health-and-care-information>

Information about NHS England and the Data Access Request Service can be found here:

<https://digital.nhs.uk/services/data-access-request-service-dars/how-nhs-digital-makes-decisions-about-data-access>

<https://digital.nhs.uk/services/data-access-request-service-dars#further-information>

### **Wales:**

SAILS is a Trusted Research Environment (TRE), which is a safe, secure, computing environment where researchers, scientists and other experts can gain access to data for research in the public interest, via a rigorous approval process. It is funded by Health and Care Research Wales through the Welsh government.

The Information Governance Review Panel (IGRP) provides independent guidance and advice on Information Governance policies, procedures and processes for SAIL Databank. All applications to use the SAIL database are overseen by the Information Governance review panel (IGRP)

Information about data management policies within SAIL can be found here:

<https://saildatabank.com/governance/approvals-public-engagement/information-governance/>

<https://saildatabank.com/about-us/policies/>

### **Scotland:**

As mentioned above, **NHS Scotland** Public Benefit and Privacy Panel for Health and Social Care (**HSC-PBPP**) is a governance structure of NHS Scotland (NHSS) who scrutinise and approve applications to health data. Data access is managed and overseen by the electronic Data Research and Innovation Service (**eDRIS**), whose team are part of Public Health Scotland.

Information about the governance role of HSC-PBPP can be found here:

<https://www.informationgovernance.scot.nhs.uk/pbpphsc/>

The privacy policy of Public Health Scotland can be found here:



<https://www.publichealthscotland.scot/our-privacy-notice/organisational-background/>

### **Northern Ireland:**

Within Health and Social Care Northern Ireland (HSCNI), the Honest Broker Service (HBS) is responsible for Information Governance and access to anonymised data for ethically approved research.

Secure access to de-identified data via a safe setting is via the Health Data Research Northern Ireland UK Secure e-Research Platform (HDRNI UK SeRP) which was established in March 2021.

Further information about the privacy policy of the Honest Broker Service can be found here:

[https://hscbusiness.hscni.net/pdf/HBS\\_Privacy\\_Notice\\_Feb21.pdf](https://hscbusiness.hscni.net/pdf/HBS_Privacy_Notice_Feb21.pdf)

### *49. What will my data be used for?*

Examples of some research that can be conducted using your questionnaire data or medical records are:

- Identifying which physical and mental conditions are more common in autistic individuals compared to their non-autistic family members
- Understanding what factors contribute to self-reported quality of life in autistic individuals
- Investigating if camouflaging and masking contribute to depression and anxiety in autistic individuals

### *50. How will the study account for possible inaccuracy in medical records?*

At registration, participants will also be asked to complete a questionnaire about health conditions they have been diagnosed with, or suspected undiagnosed health conditions. By using both health-related information provided by participants or parents/carers of participating children alongside information on health from electronic health records, we can account for potential inaccuracy in medical records.

### *51. Is sharing medical records compulsory?*

We are open to discussion with the consultees about making accessing electronic health records optional.

## *I. Additional ethical questions based on inclusion of children*

### *52. What is the consent procedure for including children in Spectrum 10K (including withdrawal and re-consent)?*

For children up to the age of 16, a parent or a legal guardian will need to provide consent for the child to participate in Spectrum 10K.

For children 6 years of age or over, and who do not have a diagnosis of intellectual disability and can read and understand information appropriate for their age, we will additionally provide adapted information sheets. These documents will provide the child with information about the study in an easier read format and explain what is involved in taking part. Following research ethics guidelines, we will ask that the child completes an 'assent form' which asks 'yes' or 'no' questions to confirm they are happy to participate. This is not obligatory, but we recommend that this step is completed.

When the child turns 16 years of age during the study period, we will contact the parent/legal guardian to invite their child to provide their own consent to continue being part of Spectrum 10K. This would allow the 16-year-old to be invited to further studies.

Please note that this aspect of the study is currently being discussed with the HRA. The Spectrum 10K team will update consultees when more information becomes available.

### *53. What is meant by 'assent'?*

Children who are under the age of 16 are not legally capable of providing consent. However, children who are able to read are able to provide 'assent' or agreement.

For any children who are at least 6 years old, who do not have a diagnosis of intellectual disability, and whose parents indicate that they are able to read and understand information appropriate for their age, we will seek their assent to participate in Spectrum 10K.

To do this, we will provide the child's parent with documents about Spectrum 10K that have been adapted to be easily read by children of the same age as their child. We will then request that the parent ask their child to read these documents and complete an 'assent form'. The assent form will contain simple 'yes or no' questions about whether they have understood the study and are happy to take part.

We will not register as participants children who indicate that they do not assent to taking part in Spectrum 10K, regardless of whether or not their parent has consented on their behalf. We will only register children if either:



- their parent consents on their behalf **and** the child provides assent, or
- their parent consents on their behalf **and** the child does not complete the assent form.

This procedure is in line with independent research ethics guidelines detailed in the following document: <https://www.hra.nhs.uk/planning-and-improving-research/policies-standards-legislation/research-involving-children/>

#### *54. What will happen to DNA once a child becomes an adult?*

Once a child turns 16 years of age, as is standard practice, their data (including DNA data) is stored and kept for the purpose of the study, as consented for by their parent(s).

Children who turn 16 have the option to withdraw themselves from Spectrum 10K. However, any data collected up to that point will be kept for archival purposes only. This means that their data, including DNA, cannot be used in future research, and is only stored to verify the validity of previous analysis.

#### *55. Is it ethical to use DNA from children based on consent from their parents/carers, given that this cannot be retracted if the child decides later that they do not consent?*

Easy-to-read and age-appropriate information sheets that give children the opportunity to understand and agree are typical of research studies that involve children.

Whilst children under the age of 16 years cannot legally provide consent, children who can read can provide assent (agreement). An assent form is available for children in addition to a parent consent form.

Spectrum 10K has created easy-to-read, age-appropriate information sheets that provide an explanation of the study. Children can choose to be enrolled in the study only if they agree.

For children who are six and above, you can find these at:

<https://Spectrum10K.org/information-sheets/>.

For dependent adults who are able to read, there is an easy-to-read consent form.

For adults who lack the capacity to consent to participate in Spectrum 10K, a consultee is required and they can access a consultee information sheet and consultee declaration form.



These study procedures have been assessed by a specialist ethics committee with expertise in research studies involving adults who lack capacity to consent to participate in research studies.

When children turn 16, we will contact the parents to ask their child to consent in their own right if they have the capacity to consent. They can choose to withdraw from Spectrum 10K at this time. Our data maintenance plan will ensure that withdrawals from Spectrum 10K are possible for as long as the dataset is stored.

This plan is based on existing and long-running research studies that involve young people such as ALSPAC and Born in Bradford.

### *J. Inclusion of adults without capacity to consent to participate in the study*

*56. Why is it important to include adults without capacity to consent to participate?*

*57. How does Spectrum 10K include adults without capacity to consent to participate?*

*58. How has participation of autistic individuals with limited communication to participate been designed? How has this been vetted?*

*59. How can capacity to consent to participate in Spectrum 10K be assessed?*

*60. Is it ethical to use DNA from adults who lack the capacity to consent to participate, given that this cannot be retracted if the person is ever able to review the decision for themselves?*

We are currently in discussions with the HRA to explore this aspect of the study and will be updating consultation participants once we know more.





## *K. Ambassadors*

### *61. What is the ongoing role of Ambassadors in Spectrum 10K?*

Ambassadors were non-paid members of the autism community who were interested in the study and in promoting it. As of 23<sup>rd</sup> May 2022 we have decided to end the role of Ambassadors for the study. We are very grateful to our Ambassadors for the support they have provided, but it will be impossible to keep Ambassadors up to date as the consultation progresses because things can change very quickly. This risks placing Ambassadors in a situation where they are asked questions that they cannot answer, simply because we have not had time to bring them up to date. We want to avoid any risk of public confusion about the project and focus entirely on the consultation.

## *L. Autism community involvement prior to pause*

### *62. How were autistic people involved in the design and setup of Spectrum 10K?*

No autistic people were involved in the design of Spectrum 10K prior to the Wellcome Trust providing funding for the project. After Spectrum 10K was funded, we recruited an Advisory Panel made up of the following people:

- 3 autistic adults.
- 3 non-autistic parents/carers of autistic children.
- 2 clinicians (clinical psychologists/psychiatrists).
- 1 representative from the autism-related charity Autistica.

We asked our Advisory Panel to do the following:

Tell us what they thought were the most important areas of research for autistic people. Together, we agreed that these areas included mental and physical health conditions, education, social relationships, diet, sleep, employment, and income.

Review the list of questionnaires that participants in Spectrum 10K would be asked to complete and suggest changes. Based on their feedback, we added whole new questionnaires about vulnerability and quality of life. Together, we also agreed that it would be important to ask questions about autistic and sensory traits, anxiety, depression, obsessive compulsive disorder, developmental milestones, and camouflaging (or masking).



Review public-facing information, including the information sheets, that we would provide participants in Spectrum 10K. Based on their feedback, we made some changes to the information to make sure that they were clear.

We incorporated the feedback of our Advisory Panel into the creation of the Spectrum 10K study protocol, which is the manual that the research team will follow as they carry out the study.

### *63. Why were relatives and other stakeholders involved?*

We invited parents, psychologists working with autistic people, and an autism charity representative to take part in the Advisory Panel to represent the voices of autistic children as well as to gain insight from those who work closely with autistic people and have an interest in their wellbeing.

### *64. What are you going to do to engage the autism community more widely?*

We have heard the feedback from the wider autism community online and we recognise that we need to consult with more and diverse autistic people and carers.

In response to the feedback since our launch we commissioned Hopkins Van Mil to work with us on a Spectrum 10K consultation. As a result we have worked with autistic people and carers to co-design a consultation strategy and begin a wider consultation process.

The Spectrum 10K consultation is being co-led by Hopkins Van Mil, which is an independent agency, by Leneh Buckle, who is an autistic researcher working on a freelance basis for Hopkins Van Mil, and the Spectrum 10K team.

Some of what we will be asking for feedback on includes:

- How to make information about the study as clear as possible
- How to make the study safer, more ethical, or improved in all aspects of its work
- What specific issues do autistic people want us to research
- How should our data access request process work so autistic people and families feel comfortable sharing
- Who should be on the Data Access Committee
- What criteria should the Data Access Committee use to decide whether to share data
- How we should manage data sharing after the project has ended

This feedback can be provided in:

- A consultation survey
- A series of online webinars and discussion groups



- One to one interviews
- Consultation activities will be open to everyone who wishes to contribute to the improvement of Spectrum 10K: with autistic people being the main consultees, with their families, researchers, and health and social care professionals.

These activities will provide opportunities to hear from the diverse voices of the autism community so that we can collaborate and shape Spectrum 10K together.

We want to ensure that the autism community is meaningfully involved in this project.

The Spectrum 10K team will continue to consult the autism community, being transparent with our process and our goals, to ensure we can work together throughout the length of the project.

### *65. What issues will be covered when you engage the autism community more widely?*

In response to the feedback since our launch, we have co-designed a consultation strategy with autistic people.

Some of what we will be consulting about might include:

- How to make information about the study as clear as possible
- How to make the study safer, more ethical, or improved in all aspects of its work
- How should our data access request process work so autistic people and families feel comfortable sharing their data (genetic, questionnaire, and linked to electronic health records)?
- Who should be on the Data Access Committee?
- What criteria should the Data Access Committee use to decide whether to share data with external researchers?
- How should we manage data sharing after Spectrum 10K has ended?

### *M. Next Steps*

### *66. When will the study restart? And how long will it take to complete?*

This will depend on when the consultation ends and how long changes to the project will take to complete. At the current time, the consultation is starting in February 2023 and is expected to end in April 2023. A working group is expected to convene in April 2023 to implement the changes into the study, study documentation and website. Following submission of study changes and approval from our Research Ethics Committee and Health Research Authority, we hope to



restart the study in August 2023, although we are mindful that delays may be incurred. Following the re-launch of Spectrum 10K it is expected that participant recruitment will resume and data collection will occur over a 24 month period, subject to approval from the study funder.