



This is what Spectrum 10K has said about:

## *The aims of the study*

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

#### 1.1 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.

#### 1.2 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.

#### 1.3 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.