



This is what Spectrum 10K has said about:

Development of a prenatal test

1. 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.

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.

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.