Great Ormond Street Hospital for Children
NHS Foundation Trust

Genetics of Autistic Spectrum Disorders: information for families

This information sheet from Great Ormond Street Hospital (GOSH) Clinical Genetics Department explains what we know about the genetic background to autistic spectrum disorders. It accompanies our general information sheet on autistic spectrum disorders, available on the GOSH website.

What is an autistic spectrum disorder?

The diagnosis of autism is complex and based on observation of a child's developmental progress and behaviour over time by one or more specialists working within a Community Paediatric team. Autism is now regarded as a 'spectrum' disorder as the symptoms can vary from mild to severe. There is also variation in symptom severity within families.

For instance, some children with autism may have a significant intellectual disability, while others have high intelligence.

Is ASD common?

ASD is common and affects around 11 in every 1,000 people, which is 1.1 per cent of the population. This means that 700,000 people in the UK are thought to have ASD. Through increased awareness among parents and health professionals, the diagnosis is now made more frequently than previously.

Is ASD more common in boys or girls?

ASD is approximately four times more commonly seen in boys than girls, but the reasons for this difference are poorly understood.

What causes ASD?

The exact cause of autism is not known, and it is likely that there are different causes affecting different families. Research has pointed to several possible contributing factors, including genetic (heredity) and non-genetic (environmental) factors.

Up to 10 per cent of ASD cases occur in children who also have another condition of known genetic origin (such as Fragile X syndrome or tuberous sclerosis). As part of their paediatric assessment, your child should have been examined to look for clinical features of these conditions. They should also have had a blood test to check for Fragile X syndrome in addition to detectable chromosome abnormalities.

For the remaining 90 per cent or more of individuals with ASD, the causes are not yet known.

We think that there is a significant genetic component. Studies of twins have shown it is more likely that both twins will be affected when they are genetically identical. Studies of relatives of an individual with ASD show that they have a higher chance of being affected than unrelated people in the general population.

In some families, there appear to be genetic risk factors which have been passed down from one generation to the next. In others, the genetic risk factors appear to have started as a completely new event, just by chance (sporadically), when the genes and chromosomes have been copied to pass on in the egg or sperm of the parent. In other families, we believe that the affected individual has changes in several different genes, each of which carries a small increased risk, but which become significant when added together.

We also know from studies of identical twins that sometimes only one twin is affected. This suggests that there may also be non-genetic factors contributing. These 'environmental' causes are not usually possible to identify at present.

What is the chance that a younger brother or sister of a child with ASD will also be affected?

It is very difficult to accurately estimate the chance that a younger brother or sister of a child with ASD will also fit broadly within an autism spectrum diagnosis. Our current estimate lies somewhere between 10 and 20 per cent based on research studies to date. Given the relative frequency in males, the chance that a younger brother will develop ASD is higher than the chance that a younger sister will develop ASD. The risk of a brother or sister developing the more general symptoms of ASD, which fall towards one end of the normal behavioural spectrum, including speech delay, poor social language abilities and social reserve, may be up to 30 per cent.

Can I my baby be tested for autism in the womb?

Unless a specific genetic diagnosis has been identified, such as tuberous sclerosis or Fragile X syndrome, there is no test available which can predict whether the baby being carried will develop autism. This is the situation for the vast majority of families. Specific prenatal diagnostic tests, including copy number variants (CNVs) of uncertain significance or fetal sexing for males, are not sensitive or accurate enough for use in clinical practice.

Do I need to see a geneticist if I want more information about having further children?

Currently geneticists can offer little additional advice to families where there is an individual with ASD, other than provide general non-specific risk information outlined above. If you believe that your child has not had routine tests, including a detailed chromosome test known as an 'array CGH' and a Fragile X test, please discuss this with your Community Paediatrician.

If your child has unusual physical characteristics, severe epilepsy or you have two or more affected children, a genetics referral may be indicated and we suggest that you discuss this with your Community Paediatrician.

Is our understanding likely to change in the future?

Research to date suggests that there may be very many different causes of ASD operating in different families. For most families, it is unlikely that we will be able to identify the specific causes in their child with ASD in the near future. That means it is unlikely we will be able to offer testing for ASD during pregnancy in the near future.

However, there are many large research studies into the causes of ASD taking place across the world. We hope that as genetic and non-genetic risk factors are identified, and we begin to understand how they work together, we will develop a better understanding of ASD which will in turn lead to improved treatment, prevention and assessment of recurrence risks. Significant advances are likely to be reported in national media, through the ASD family support groups and updates to paediatricians and other specialists in the field.

Further information and support

The National Autistic Society is the main organisation in the UK offering support and advice to anyone affected by an ASD. Call their helpline on 0808 808 4104 (Monday to Thursday from 10am to 4pm and Fridays from 9am to 3pm). You can also visit their website at www.autism.org.uk

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