Is autism genetically inherited?

The idea that autism is, at least in part, genetically determined is strongly supported by current research. The exact ways that genetics influences the likelihood that a person is on the autism spectrum are still to be determined by researchers. So far, no individual gene, set of genes or genetic variation has been categorically identified as determining autism in an individual.1

Comparing twins who are on the autism spectrum

One of the most reliable methods for assessing the heritability of a condition is to conduct a twin study, which compares a sample group of identical twins and another of non-identical (fraternal) twin, and records whether one, both or neither of the twins in each sibling pair has that condition. Because identical twins have exactly the same genetic profile, any genetically determined condition that affects one twin is almost certain to affect the other as well. On the other hand, non-identical twins (fraternal twins) have only around half of their genetic material in common, so there is a much smaller chance that both non-identical twins in a pair will inherit a genetically determined condition.

In addition, because most pairs of twins grow up in the same household, they are likely to share very similar experiences relating to the home, family life and upbringing, such as same parenting styles, play opportunities, family activities, and schooling.

If autism could be traced solely to external factors and genetics played no part at all, then we would expect to see similar concordance rates for autism in both identical and non-identical twins.

However, researchers have found that if one of a pair of fraternal twin siblings is on the autism spectrum, there is 20% chance that the second twin is also on the autism spectrum, whereas if one of a pair of identical twins is on the autism spectrum, there is approximately a 90% chance that the second twin is also on the autism spectrum.2,3
**Autism in families**

Research into autism in families has also found that:

- Individual autism traits, such as obsessive behaviour, communication deficits and impaired social interaction, are highly heritable. ⁴ ⁵
- Autism is often present in multiple cases within extended families (called 'multiplex' families) and sometimes only one family member is on the autism spectrum (called 'simplex' families).
- Some of the cases where only one family member is on the autism spectrum may be due to isolated, spontaneous changes to DNA sequences that occur at the cell formation stage. It is believed that these rare mutations account for around 10% of all people on the autism spectrum.⁶
- In families where more than one individual is on the autism spectrum, and/or in which several members have strong autism traits, it is thought that specific genetic variations, passed down through generations, might underlie the increased heritability for autism.¹
- Learned behaviours within the family environment may also play a role in the inheritability of autism traits. For example, a 2017 study has reported that children whose parents have a neurodevelopmental condition such as autism tend to also have severe restricted interest and repetitive behaviours (a core trait of autism) even if the child does not have a diagnosis of autism.⁷

**Genetic markers for autism**

To date, over 700 genetic loci and 61 genes have been identified as being possibly associated with autism.⁸ However, as more genes and loci are identified, it is becoming clear that the genomic architecture of autism is complex and highly heterogeneous. Researchers have found that some individuals have multiple genetic variations. These changes can interact with each other and may explain the varied experiences of people on the spectrum.

A recent study suggests that mutations in genes give rise to autism’s behavioural traits, whereas large DNA duplications or deletions, called copy number variations (CNVs) are associated with the thinking style of people on the autism spectrum.⁸

Identifying genetic markers for autism may provide options for early diagnosis and interventions. Researchers have recently developed a blood test that may possibly identify differences in brain cell development in children as young as one year of age.⁹
Australia’s first and largest autism Biobank – you can help

Australia’s Cooperative Research Centre for Living with Autism (Autism CRC) is creating Australia’s first and largest autism biobank. The Biobank aims to collect detailed biological information from over 1200 Australian children on the autism spectrum and their families. Aspect is a key partner in the Autism CRC, a national research program designed to improve the lives of people on the autism spectrum and their families across the lifespan.

By understanding how genes and gene combinations may result in different types of autism, the Autism CRC is seeking to learn much more about why and how autism occurs. It is hoped that this will lead to a genetic discovery that can enable earlier and more accurate diagnosis.

To find out more about the Biobank and how to take part, go to http://www.autismcrc.com.au/autism-crc-biobank-participant-information

References


