Familial risk of autism spectrum disorder

What is the issue?

Familial risk (or ‘heritability’) refers to the extent to which a trait or condition is genetically determined, as opposed to being triggered by environmental factors. Given that no single ‘cause’ for autism spectrum disorder (ASD) has yet been pinpointed, the role of genetics in the development of these conditions remains a matter of some debate.

This issue may be of concern to adults who have a diagnosis ASD and who want to understand the likelihood of their children ‘inheriting’ the condition. It may also be significant for parents who already have a child on the autism spectrum and who are unsure whether any subsequent children they have may likewise be born with ASD.

What does the research say?

One of the most reliable methods for assessing the heritability of a condition is the twin study. To conduct a twin study, researchers must gather a sample group of identical twins and another of non-identical (fraternal) twins. They will then record whether an ASD is present in one, both or neither of the twins in each sibling pair.

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 – in other words, it will have a close to 100 per cent concordance rate. On the other hand, non-identical twins have only around half of their genetic material in common. There is therefore a much smaller chance that both non-identical twins in a pair will inherit a genetically determined condition.

Twin studies enable researchers to make judgements about the role of environmental factors in determining developmental outcomes. Since 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. For example, they will probably be exposed to the same parenting styles, play opportunities, family activities, and schooling. If ASD could be traced solely to external factors such as these (and genetics played no part at all) then we would expect to see similar concordance rates for the condition between both identical and non-identical twins.

Researchers have found that if one of a pair of identical twins meets the diagnostic criteria for ASD there is approximately a 90 per cent chance that the second twin will also have the disorder. In contrast, the concordance rate for ASD between fraternal twin siblings seems to around 10 per cent (Bailey et al., 1995).

The idea that ASD is, at least in part, genetically determined has been further supported by research indicating that ASD is often present in multiple cases within extended families (Silverman et al., 2002; Lauritsen et al., 2005). Additionally, it has been found
that individual ASD traits, such as obsessive behaviour, communication deficits and impaired social interaction, are highly heritable (Ronald et al., 2005; Zhao et al., 2007).

Research into families who have members with ASD have shown that:

- There are some families where only one member has a diagnosis of ASD. These 'one-off' incidences are referred to as 'simplex' ASD. Genetics research suggests that some of these cases 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 one in ten of all people diagnosed with ASD (Sebat et al., 2007).
- On the other hand, there are many families in which more than one individual has an ASD diagnosis, and/or in which several members have strong ASD traits. These are referred to as 'multiplex' families. It is thought that there are specific genetic variations, passed down through generations, which might underlie the increased familial risk for ASD in these cases. However, there is no evidence as yet to prove that the condition is traceable to a single, consistent genetic defect.

Identifying individuals who have genetic ASD markers may in the future provide options for early diagnosis and interventions. Researchers have recently developed a blood test that looks promising for identifying differences in brain cell development in children as young as one year of age (Courchesne, 2013). As the multi-genetic basis of ASD is discovered new diagnostic and treatment options should follow.

In summary

The results of twin and family studies to date indicate that there is almost certainly a genetic component to ASD. Whilst there appears to be a growing body of evidence to suggest that particular genetic variations, passed through families, may predispose some individuals to a greater likelihood of developing ASD, at this point in time no individual gene or set of genes has been categorically identified.

References


**Resources**

What support is available? Australian Government resources