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|  | What causes Autism Spectrum Disorder (ASD)? |
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ASD has no single known cause. Given its complexity and the fact that symptoms and severity vary, there are probably many causes including a range of genetic and environmental risk factors.

Genes and environment

Theories about what might cause ASD have changed over the past sixty years. In some families, there appears to be a pattern of ASD or other disabilities. It also appears that some children are born with a susceptibility to autism, but researchers have not yet identified a single “trigger” that causes autism to develop1.

Evidence supporting a significant genetic contribution to ASD has been increasing over the past few decades, but what causes or increases the risk for ASD is not yet understood2.

Studies have shown that among identical twins, if one child has an ASD, then the other will be affected between 36-95% of the time. In non-identical twins, if one child has an ASD, then the other is affected between 0-31% of the time.

The patterns of genetic inheritance are complex, with the involvement of multiple genes, rather than a simple dominant, recessive or X-linked gene2. A number of chromosome regions are likely to contain risk genes for ASD, and no gene has been unequivocally identified to date3.

Research has begun to show that environmental factors may also involve some genetic susceptibility. A recent study shows that at an individual level, the risk of autism increases according to how close you are genetically to other relatives with autism4.

Other possible causes

About 10% of people with an ASD have known medical conditions, including Fragile X syndrome, tuberous sclerosis, congenital rubella syndrome, and untreated phenylketonuria (PKU). A small percentage of children who are born prematurely or with low birth weight are at greater risk of ASD. Advanced paternal and maternal age, growth restriction and newborn hypoxia are also associated with an increased risk of ASD.5, 6, 7

ASD and immunisation – no link

Over the past decade there has been considerable discussion, publicity and research concerning whether there is a link between autism and immunisation. Comprehensive studies have provided no evidence to support a link between measles-mumps-rubella (MMR) vaccination and ASD. Previous studies suggesting a causal link were found to be seriously flawed. There is also no evidence to suggest that any other childhood vaccine may increase the risk of ASD and no association between the use of preservatives such as thimerosal that contains ethyl mercury in vaccines and ASD.8

What is the risk of ASD if you want to have other children?

In the general population, ASD affects about 1 in 100 children9. A recent study provided measures of individual risk for children who have a relative with an ASD.

This study identified that children with a sibling with an ASD are ten times more likely to develop ASD; three times more likely if they have a half-brother or sister with an ASD; and two times more likely if they have a cousin with an ASD. There are no differences in relative risk between genders4.

References

1. Constantino, J. N., et al. "Autism recurrence in half siblings: strong support for genetic mechanisms of transmission in ASD." Mol Psychiatry 18.2 (2013): 137-138.

2. Gupta, A. R., and State, M.W. (2007). Recent Advances in the Genetics of Autism. Biological Psychiatry, 61, 429-437.

3. Yang, M. S., and Gill, M. (2007). A review of gene linkage, association and expression studies in autism and an assessment of convergent evidence. International Journal of Developmental Neuroscience, 25(2), 69-85.

4. Sandin,S., Lichtenstein,P., Kuja-Halkola, R., Larsson, H., Hultman, C., and Reichenberg, A. (2014). The Familial Risk of Autism. JAMA, 311 (17): 1770

5. Kolevzon, A., Gross, R., and Reichenberg, A. (2007). Prenatal and Perinatal risk factors for autism. Archive Pediatric Adolescent Medicine, 161, 326-333.

6. Lundström, S., Hamworth, C., Carlström, E., Gillberg, C., Mill, J., Rastam, C., Reichenberg, A. (2010). Trajectories leading to autism spectrum disorders are affected by paternal age: findings from two nationally representative twin studies. Journal of Child Psychology and Psychiatry, 51(7), 850-856.

7. Gardener, H., Spiegelman, D., and Buka, S. (2011). Perinatal and Neonatal Risk Factors for Autism: A Comprehensive Meta-analysis. Pediatrics, 128(2), 344-355.

8. Fombonne, E. (2008). Thimerosal disappears but autism remains. Archive of General Psychiatry (65), 15–16.

9. Elsabbagh, M., Divan, G., Koh, Y.-J., Kim, Y. S., Kauchali, S., Marcín, C., Montiel-Nava, C., Patel, V., Paula, C. S., Wang, C., Yasamy, M. T. and Fombonne, E. (2012), Global Prevalence of Autism and Other Pervasive Developmental Disorders. Autism Res, 5: 160–179.

For more information about the Department of Education and Early Childhood Development’s Autism Friendly Learning website go to:

[www.education.vic.gov.au/autism](http://www.education.vic.gov.au/autism)