

Genetics & Autism

Efficacy Rating
NA

What are the issues?

Current research on the genetics and autism suggests that up to 5-10% of autism is secondary to a chromosome abnormality or a single gene disorder. (Muhle et al, 2004) The remaining 90-95% of cases are of unknown cause yet further advances in genetics may determine other single gene disorders not yet detectable.

Around 30% of children with autism have dysmorphic features such as microcephaly and/or brain abnormalities, while 70% have no physical abnormalities. Recurrence risks for autism appear to be different in subsequently born siblings based on the gender of the sibling, and not the gender of the sibling prior diagnosed with autism. (Newshaffer et al, 2002)

Recurrence risks also differ based on presence or absence of physical features in the prior diagnosed child.

The reader is cautioned however that these and other cited recurrence risks are often from small studies and should not be used in absolute terms for genetic counseling purposes.

For male siblings of an affected child the risk are about 7 % if the affected child has no physical manifestations and about 1% if the affected child has physical differences. (Ritvo et al, 1989) For a female sibling of an affected child the risks are about 1% in either scenario. (Ritvo et al, 1989)

The likelihood of autism increases dramatically toward 35% if there are two previously affected siblings. (Ritvo et al, 1989)

Muhle, R., Trentacoste, S.V., & Rapin, I. (2004). The genetics of autism. *Pediatrics*, 13, 472-486.

Genetics & Autism Resources and Information

Articles:

Muhle, R., Trentacoste, S.V., & Rapin, I. (2004). The genetics of autism. *Pediatrics*, 113, e472-486.

Newschaffer, C.J., Fallin, D., & Lee, N.L. (2002). Heritable and Nonheritable Risk Factors for Autism Spectrum Disorders *Epidemiologic Reviews*, 24, No. 2.

Ritvo, E.R., Jorde, L.B., Mason-Brothers, A., Freeman, B.J., Pingree, C., Jones, M.B., McMahon, W.M., Petersen, P.B., Jenson, W.R., Mo, A. (1989). The UCLA-University of Utah epidemiologic survey of autism: recurrence risk estimates and genetic counseling. *Am J Psychiatry*, 146, 1032-1036.

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