

Genetic factors may explain most of risk for autism spectrum disorder

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Quinn, an autistic boy, and the line of toys he made before falling asleep. Repeatedly stacking or lining up objects is a behavior commonly associated with autism. Credit: Wikipedia.

Reanalysis of data from a previous study on the familial risk of autism spectrum disorder (ASD) estimates the heritability to be 83 percent, suggesting that genetic factors may explain most of the risk for ASD, according to a study published by *JAMA*.

Studies have found that [autism spectrum disorder](#) (ASD) aggregates in families. In a previous study, ASD heritability was estimated to be 50 percent. To define presence or absence of ASD, the study used a data set created to take into account time-to-event effects in the data, which may have reduced the heritability estimates. Using the same underlying data from this study, Sven Sandin, Ph.D., of the Icahn School of Medicine at Mount Sinai, New York, and colleagues used an alternate method (used by previous studies in the field) to calculate the heritability of ASD.

The study included a group of children born in Sweden 1982 through 2006, with follow-up for ASD through December 2009. The analysis included 37,570 [twin pairs](#), 2,642,064 full sibling pairs, and 432,281 maternal and 445,531 paternal half-sibling pairs. Of these, 14,516 children were diagnosed with ASD. Various models were tested and using the best-fitting model, the ASD heritability was estimated as 83 percent and the nonshared environmental influence was estimated as 17 percent.

"This estimate [83 percent] is slightly lower than the approximately 90 percent estimate reported in earlier twin studies and higher than the 38 percent estimate reported in a California twin study, but was estimated with higher precision. Like earlier twin studies, shared environmental factors contributed minimally to the risk of ASD," the authors write.

The researchers note that twin and family methods for calculating heritability require several, often untestable assumptions. Because ASD is rare, estimates of heritability rely on few families with more than one affected child, and, coupled with the time trends in ASD prevalence, the heritability estimates are sensitive to the choice of methods.

"The [method](#) initially chosen in the previous study led to a lower estimate of heritability of ASD. The current estimate, using traditional methods for defining ASD discordance and concordance, more

accurately captures the role of the [genetic factors](#) in ASD. However, in both analyses, the [heritability](#) of ASD was high and the risk of ASD increased with increasing genetic relatedness," the researchers write.

More information: *JAMA* (2017). [DOI: 10.1001/jama.2017.12141](https://doi.org/10.1001/jama.2017.12141)

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