⭐ Autism spectrum disorder (Matoba, 2020)
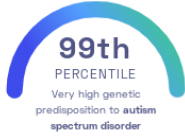
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Molecular Psychiatry

`Brain`

STUDY SUMMARY

Discovery of a novel genomic region associated with autism spectrum disorder (ASD).

YOUR RESULT

99th
PERCENTILE
Very high genetic
predisposition to **autism
spectrum disorder**

STUDY DESCRIPTION

Autism spectrum disorder (ASD) is a condition that affects the brain's development. It impacts social skills, speech, and learning. ASD is a highly heritable condition, yet previous studies have identified only a small number of genetic factors. This study found 5 regions in the genome associated with ASD by examining the genetic data of over 6,000 individuals of European, African, and East Asian ancestries. One of these regions had not been identified by any previous study. Interestingly, this newly-identified region has previously been connected to schizophrenia, bipolar disorder, and obsessive-compulsive disorder. The study further showed that genetic variation in this region may affect a gene called DDHD2, which plays a role in moving molecular cargo inside cells.

DID YOU KNOW?

Symptoms of autism appear differently in males and females, and as result females with the condition often take longer to be diagnosed.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to autism spectrum disorder we summed up the effects of genetic variants that were linked to autism spectrum disorder in the study that this report is based on. These variants can be found in the table below. The variants highlighted in green have **positive effect sizes** and increase your genetic predisposition to autism spectrum disorder. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to autism spectrum disorder. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to autism spectrum disorder. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for autism spectrum disorder to be 0.34**. To determine whether your score is high or low, we compared it to the scores of 5,000 other Nebula Genomics users. We found that your polygenic score for autism spectrum disorder is in the **99th percentile**. This means that it is higher than the polygenic scores 99% of people. We consider this to be a **very high genetic predisposition to autism spectrum disorder**. However, please note that genetic predispositions do not account for important non-genetic factors like lifestyle. Furthermore, the genetics of most traits has not been fully understood yet and many associations between traits and genetic variants remain unknown. For additional explanations, click on the column titles in the table below and visit our Nebula Library tutorial.

VARIANT ⓘ	YOUR GENOTYPE ⓘ	EFFECT SIZE ⓘ	VARIANT FREQUENCY ⓘ	SIGNIFICANCE ⓘ
rs1000177_T	T / T	0.09 (↑)	24%	1.34×10^{-9}
rs716219_T	C / C	0.08 (−)	34%	6.42×10^{-9}
rs10099100_C	G / G	0.08 (−)	31%	7.65×10^{-9}
rs112436750_A	A / A	0.09 (↑)	21%	2.62×10^{-8}
rs60527016_C 🏅NEW	T / T	-0.07 (−)	21%	4.70×10^{-8}