

# Genome testing for siblings of kids with autism may detect ASD before symptoms appear

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One of the key priorities of interventions for autism spectrum disorder (ASD) is starting early, with some evidence showing infants as young as seven months old could benefit. Yet, most children in North America aren't diagnosed with ASD until they're over four years of age. New research led by The Hospital for Sick Children (SickKids) and the University of Alberta published on December 5, 2019 in *Nature Communications* has found testing the DNA of siblings of individuals with ASD may be predictive of a future diagnosis even if symptoms aren't yet apparent.

ASD refers to a group of neurodevelopmental conditions resulting in challenges related to communication, social understanding and behaviour. Studies show families who have a child with ASD have a 6.9 to 19.5 per cent chance of another child having ASD and a 30 to 40 per cent chance of another child having atypical development.

## **Genomic factors linked to ASD-related traits**

"Genetic factors are the most likely reason we see a clustering of ASD related traits in families," explains Dr. Stephen Scherer, Senior Scientist and Director of The Centre for Applied Genomics (TCAG) at SickKids, Director of the McLaughlin Centre at the University of Toronto and principal investigator of the study. "We wanted to investigate the possible benefits of genetic testing for infants whose older sibling had already been diagnosed with ASD. If we can identify those children early, we may be able to enrol them earlier in therapies."

The researchers looked for the presence of genetic alterations that have been linked to ASD called copy number variations (CNVs) in over 288

infant siblings from 253 families. By age 3, 157 siblings were either diagnosed with ASD or developing atypically. DNA testing revealed CNVs in genes relevant to ASD in 11 (7 per cent) of the 157 siblings who were eventually diagnosed.

The study found that the presence of an ASD-relevant CNV in a [sibling](#) had a high likelihood of predicting a future diagnosis of ASD or atypical development. This marks the first time that scientists have been able to quantify the predictive value of CNVs in determining these diagnoses.

## **Early identification could lead to earlier intervention**

"These findings add to a growing body of evidence that biomarkers might be helpful in identifying pre-symptomatic infants who are likely to develop ASD or other developmental challenges," says Dr. Lonnie Zwaigenbaum, Professor of Pediatrics, Stollery Children's Hospital Foundation Chair in Autism and Stollery Science Lab Distinguished Researcher at the University of Alberta.

"At this point, we can't fully determine the anticipated severity of a child's future symptoms. What we can say is that it's important to closely monitor their development and start therapeutic interventions early to support their skill development and address emerging functional impairments related to ASD."

The research team has confirmed similar findings in a separate group of 2,110 families having one child with, and a second child without ASD. Their next step will be to look beyond CNVs and determine how newer technologies—like whole genome sequencing—might increase the early genetic detection rate.

**More information:** L. D'Abate et al, Predictive impact of rare genomic copy number variations in siblings of individuals with autism

spectrum disorders, *Nature Communications* (2019). [DOI: 10.1038/s41467-019-13380-2](https://doi.org/10.1038/s41467-019-13380-2)

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