

Genetic mutations at conception linked to many cases of autism

Yale UniversityYale University

About 15 percent of autism cases in families with a single autistic child are associated with spontaneous mutations that occur in sex cells, Yale University researchers report in the April 4 issue of the journal *Nature*.

The findings are the result of DNA sequencing of every gene in 238 families, each with only one autistic child the most sophisticated analysis yet of the genetic links to autism. The study identified hundreds of such *de novo* or spontaneous sequence variations, and as more are discovered, medical science may find new ways to intervene in the disabling disorder, the authors say.

The Yale team, in cooperation with researchers at Carnegie Mellon University, the University of Pittsburgh and UCLA, also linked variations in three specific genes to a markedly increased risk for autism.

Prior to the advent of new DNA sequencing technology, we were largely wandering in the dark searching for autism genes, said [Matthew State](#) [1], senior author, the Donald J. Cohen Professor of Child Psychiatry, psychiatry and genetics, and co-director of the Yale Program on Neurogenetics. Now we are getting a clear view of the genetic landscape and finally have the tools in hand to find a large proportion of the many genes contributing to autism.

The genetic underpinnings of autism have been the subject of intensive research. Twin studies reveal a strong genetic component to autism but a substantial number of cases occur in families with no known history. Recent research has focused on genetic mutations that may arise at conception.

The new Yale study also showed that these *de novo* mutations were more frequent in children born to older fathers, offering at least a partial explanation for the increased risk for autism in children of older parents.

The researchers expect that, with additional DNA studies, the percentage of autism cases linked to these *de novo* mutations will increase and with it the ability to detect and treat the disorder.

With every new gene we discover, we learn more about potential treatments for patients with autism, said Stephan Sanders, pediatrician, a postdoctoral fellow in States lab and lead author of the *Nature* paper.

The study was funded by the Simons Foundation.

Other Yale researchers include Michael T. Murtha, Abha R. Gupta, John D. Murdoch,

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