

# Mutations in 3 genes linked to autism spectrum disorders

4 April 2012

Mutations in three new genes have been linked to autism, according to new studies including one with investigators at Mount Sinai School of Medicine. All three studies include lead investigators of the Autism Sequencing Consortium (ASC). The findings, in a trio of papers revealing new genetic targets in autism, are published in the April 4th online issue of the journal *Nature*. The studies provide new insights into important genetic changes and the many biological pathways that lead to autism spectrum disorders (ASD).

Gene mutations are glitches in [DNA](#) which can put you at risk for a particular disease. The genes with mutations identified in the studies - CHD8, SNC2A, and KATNAL2 - were discovered with a new state-of-the-art genomics technology known as exome sequencing, where all protein coding regions of the genome, called the exome, are analyzed. The researchers say that with further characterization of the genes and sequencing of genes in thousands of families, they will be able to develop novel therapeutics and preventive strategies for [autism](#).

"We now have a good sense of the large number of genes involved in autism and have discovered about 10 percent of them," said Joseph Buxbaum, PhD, Director of the Seaver Autism Center and Professor of Psychiatry, Genetics and Genomic Sciences, and Neuroscience at Mount Sinai School of Medicine. "We need to study many more parents and their affected children if we are to uncover the genes important in ASD. As these genes are further characterized, this will lead to earlier diagnosis and novel drug development. This work is crucial for advancing autism treatment."

In the study, ASC researchers hypothesized that de novo mutations account for a substantial fraction of the risk for autism. De novo gene mutations are mutations that show up in affected children for the first time and result from mutations in the production of sperm or egg.

Founded by Dr. Buxbaum, the Autism Sequencing Consortium is an international group of autism genetics researchers that is working to identify additional genetic causes of autism through large-scale next-generation sequencing. The institutions involved in this study sequenced data from more than 500 families (both parents and the affected child), examining the protein-enriched areas of the genome.

"When the same mutations are found in multiple affected children and none are found in children without autism, we believe that we have identified mutations that collectively affect a higher proportion of individuals with autism," said Dr. Buxbaum. "Our studies revealed that the proteins encoded by the mutated genes interact with each other far more than expected, demonstrating significantly greater connectivity than would be expected."

Two other papers from groups participating in the Autism Sequencing Consortium are also featured in the same issue of *Nature*. Led by Matthew State, PhD, Yale School of Medicine, the first identified several highly disruptive mutations in [genes](#) associated with ASD. The results show that multiple variants on one gene identify risk factors for ASD. The second study led by Evan Eichler, PhD at the University of Washington discovered that certain mutations associated with ASD are mainly of paternal origin. Their findings also support previous research showing an increased risk of developing ASD in children of older fathers.

Provided by The Mount Sinai Hospital

APA citation: Mutations in 3 genes linked to autism spectrum disorders (2012, April 4) retrieved 21 August 2022 from <https://medicalxpress.com/news/2012-04-mutations-genes-linked-autism-spectrum.html>

*This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.*