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Study adds to evidence that autism has genetic basis

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Although there is no known cause of autism, studies have shown that mutations in several genes are associated with the developmental brain disorder. New research has uncovered two additional genes that may be involved with autism.

Investigators will present their findings on Sunday, May 2 at the Pediatric Academic Societies (PAS) annual meeting in Vancouver, British Columbia, Canada.

An estimated one in 110 U.S. children has autism, which affects behavior, social skills and communication.

The risk for the disorder is higher among siblings of an affected child than in the general population, indicating that autism may be inherited, explained study co-author Ning Lei, PhD, a researcher at Princeton University and the Institute for Advanced Studies.

Dr. Lei and her colleagues analyzed data from the Autism Genetic Resource Exchange (AGRE) on 943 families, most of whom had more than one child diagnosed with autism and had undergone genetic testing. Investigators compared the prevalence of 25 gene mutations in the AGRE families with a control group of 6,317 individuals without developmental or neuropsychiatric illness.

Dr. Lei's group identified mutations in four genes within the AGRE families. Two of the genes previously were shown to be associated with

autism and often are involved in forming or maintaining neural synapses — the point of connection between individual neurons.

One of the new genes identified was neural cell adhesion molecule 2 (NCAM2). NCAM2 is expressed in the hippocampus of the human brain — a region previously associated with autism.

"While mutations in the NCAM2 gene were found in a small percentage of the children that we studied, it is fascinating that this finding continues a consistent story — that many of the genes associated with autism are involved with formation or function of the neural synapse," Dr. Lei said. "Studies such as this provide evidence that autism is a genetically based disease that affects neural connectivity."

The researchers hypothesize that a substantial percentage of children with autism will be shown to have a mutation in one or more of the many genes necessary for normal function of the synapse.

The study also showed that some parents and siblings of children with autism have the NCAM2 mutation but do not have the disorder themselves. This suggests that other environmental or genetic factors are involved in causing autism in susceptible individuals.

"These results help the public understand that autism is a very complex disorder, much like cancer," Dr. Lei said, "and no single gene or gene environment is likely to be causative in most cases."

Dr. Lei will present the study results with Daniel A. Notterman, M.D., M.A., FAAP, professor of pediatrics, biochemistry and molecular biology at Penn State College of Medicine. Dr. Notterman was associated with the faculty of Princeton University at the time these studies were performed.

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