

Researchers learn more about genetic mutation linked to autism

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University of Iowa researchers have learned more about a genetic mutation that contributes to autism. The mutation occurred in sperm cells of a father, who does not have autism, but passed the condition on to two of his children.

The investigators now know more about how the mutation causes problems with a specific gene and are testing for additional mutations of the same gene in other people with autism. Thomas Wassink, M.D., associate professor of psychiatry in the UI Carver College of Medicine, is presenting the findings May 3 at the annual International Meeting for Autism Research in Seattle.

Earlier this year, UI researchers and collaborators were part of an international team that identified, among other findings, deletions in a gene called neurexin 1, which caused the two cases of autism in one family. The UI researchers and collaborators were Wassink; Val Sheffield, M.D., Ph.D., UI professor of pediatrics and a Howard Hughes Medical Investigator; Kacie Meyer, a graduate student in Wassink's laboratory; and former UI investigator Joseph Piven, M.D., now professor of psychiatry at the University of North Carolina (UNC) and director of the UNC Neurodevelopmental Disorders Research Center,

"Genes with the most compelling evidence of causing autism appear to be components of a specific kind of neuronal connection, or synapse, called the glutamate synapse. The gene neurexin 1 was the fourth of these genes to be identified, and it is a scientifically interesting mutation

because it wasn't found in either of the parents, who do not have autism," Wassink said.

Instead, the mutation is a germline mosaic -- meaning the deletion occurred only in the father's sperm cells when he himself was in gestation. As result, the father did not have autism, but his two children, both daughters, inherited from him a chromosome that was missing a small piece of DNA that contained neurexin 1. The daughters now have autism.

Because of this missing DNA, certain proteins cannot form that normally contribute to glutamate synapses and, by extension, normal development.

"Now, using this information, we can look in a very detailed way at this gene in other families and begin to understand what happens when this protein that is normally active in the brain is missing," Wassink said.

Knowing more about how the deletions function could eventually lead to the development of diagnostic and therapeutic tools.

Source: University of Iowa

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