Researchers closer to untying autism's genetic knot

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Deciphering the functions of multiple rare genes may be at the core of understanding the genetic factors that cause autism spectrum disorders (ASDs), according to a new study published June 9 in the journal *Nature* by dozens of top autism researchers around the world, including Yale Child Study Center Director Fred R. Volkmar, M.D.

ASDs are a group of conditions marked by impairments in social interaction and communication, and by the presence of restricted and repetitive behaviors. Individuals with ASD vary greatly in cognitive development, which can range from above average to intellectual disability.

ASDs are known to be highly inheritable, but scientists are still searching for the underlying genetic determinants. Instead of focusing on a single gene responsible for ASDs, they have been looking for copy number variations (CNV), which are unusual amounts of a gene. It was once thought that people have two copies of one gene?one from the father and one from the mother?but findings have shown that the number of copies can vary. A person can have only one copy of a gene or three copies of a gene, or they can be missing the gene altogether. CNVs were once considered rare, but they are quite common. Deletions or duplications of many genes can have no effect, but other gene variations have been linked to illnesses like breast cancer or Crohn's disease.

In this study, Volkmar and his co-authors analyzed genome-wide characteristics of rare copy number variations in ASD. They compared 996 individuals with ASD of European descent to 1,287 individuals serving as controls. They found that many of these CNVs appear to be involved in the regulation of central nervous system processes. Identifying the processes related to these genes and then tracing them in animal models may lead to a better understanding of the disorder, as well as aid the search for innovative treatments.

"This paper highlights how very important genetic factors are in causing autism," said Volkmar, the Irving B. Harris Professor of Child Psychiatry, Pediatrics and Psychology Yale University School of Medicine. "It builds on earlier work which has identified several potential candidate genes and underscores that there may well be multiple genes acting to cause autism."

SOURCE [1]

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