

# Novel chromosome abnormality appears to increase risk of autism

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A multi-institutional study involving Massachusetts General Hospital (MGH) researchers has identified a chromosomal abnormality that appears to increase susceptibility to autism. In a *New England Journal of Medicine* report that is receiving early online release, the investigators – most of whom are associated with the Boston-based Autism Consortium – report that a segment of chromosome 16 is either missing or duplicated in about 1 percent of individuals with autism or related disorders, a frequency that is comparable to other genetic syndromes associated with the disorder.

“While epidemiologic studies indicate a very large genetic component to autism, little is known about how specific genes are involved,” says Mark Daly, PhD, of MGH Center for Human Genetic Research, the study’s senior author for gene discovery. “We’re still a long way from understanding how this chromosomal deletion or duplication increases the risk for autism, but this is a critical first step toward that knowledge.”

Population studies indicate that up to 90 percent of cases of autism and what are referred to as autism spectrum disorders have some genetic component, but only 10 percent of cases can be attributed to known genetic and chromosomal syndromes. Since several of those conditions involve deletions or duplications of chromosomal segments – including an inherited deletion of a region of chromosome 15 – the investigators conducted a complete genome scan of samples from the Autism Genome Research Exchange, which contains DNA from families in which at least one child has autism or a related disorder.

The scan of more than 1,400 affected individuals and a similar number of their unaffected parents revealed that an identical region of chromosome 16 was deleted in 5 individuals with an autism spectrum disorder but not in any of the parents, implying that the deletion had occurred spontaneously and was not inherited. To confirm this observation, clinical testing data from almost 1,000 patients from Children's Hospital Boston – about half of whom had been diagnosed with autism or a related developmental delay – was evaluated. Among those with a developmental disorder, 5 children had the same deletion, and in another 4 the chromosome segment was duplicated. Again, no abnormalities were seen in DNA from children without autism or developmental delay.

Confirmatory data was also obtained by colleagues from deCODE Genetics of Iceland, who found the same deletion in 3 of almost 300 individuals with an autism spectrum disorder and also in a few with other psychiatric or language disorders. Among almost 20,000 members of the deCODE database who had not been evaluated for language or psychiatric disorders, the deletion was seen in only 2 individuals. Results from the deCODE scan indicate that, while this chromosomal deletion occurs in only .01 percent of the general population, it is 100 times more prevalent in those with autism spectrum disorders.

“These large, non-inherited chromosomal deletions are extremely rare,” says Daly, “so finding precisely the same deletion in such a significant proportion of patients suggests that it is a very strong risk factor for autism. We’re now pursuing more detailed genetic studies to figure out which genes in this region are responsible for this effect in order to gain a better understanding of the underlying biology and potential clues to therapeutic approaches.”

A member of the Autism Consortium, which includes 14 Boston-area institutions, Daly adds, “The ability to rapidly and seamlessly translate

research findings to the evaluation of patients under clinical care – providing families with information that can help them understand their child’s condition and assess potential risks to other children – relied on the integrated community of researchers and clinicians made possible through the Autism Consortium.” Daly is an assistant professor of Medicine at Harvard Medical School and a member of the Broad Institute of Harvard and Massachusetts Institute of Technology.

Source: Massachusetts General Hospital

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