Scientists identify genetic link between language impairment and autism
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(Medical Xpress)—In the first molecular genetic study of families with a history of both language impairment and autism, scientists may have uncovered a shared origin for the two conditions, an important step toward explaining why some cases of autism are accompanied by language difficulties and others are not. The study, a collaboration of The Research Institute at Nationwide Children's Hospital with experts at Rutgers University, indicates that a disorder called specific language impairment—one of the most common developmental delays in children—may be caused by the same genetic variants that lead to language difficulties in some children with autism. The findings are published Oct. 30 in the American Journal of Psychiatry.

As many as two-thirds of individuals with an Autism Spectrum Disorder (ASD) also have language impairments, which can range from mild limitations to complete non-verbal behavior. However, the remaining third may have normal or even above average language abilities. To investigate whether specific language impairment and language-impaired autism cases are caused by the same genetic variants, researchers examined the genetic code of 79 families with a history of both conditions.

Using a genome-wide scan and a series of language tests, the researchers identified two new genetic links for language impairment in these families: 15q23-26 and 16p12. Each of these new links is jointly related to language-impaired ASD and non-ASD related specific language impairment, suggesting a single cause for both issues.

"A genetic cause of language impairment may help explain why some kids with ASD have language impairments and others don't, as well as why some members of a family have language impairment only and others have ASD as well," says Christopher W. Bartlett, PhD, principal investigator in the Battelle Center for Mathematical Medicine at Nationwide Children's and lead author of the study. The research is part of a long-term collaboration between scientists at Nationwide Children's and Rutgers, initiated by a grant from the National Institute of Mental Health to Linda M. Brzustowicz, MD, professor of the Department of Genetics at Rutgers and senior investigator on the project.

Language impairment is not part of the diagnostic definition of ASD. And according to Dr. Bartlett, this study raises the question of whether language impairment is actually a dissociable trait in at least some forms of ASD.

"There is nothing about autism that dictates that language impairment has to occur," says Dr. Bartlett, who also is an assistant professor of pediatrics at The Ohio State University College of Medicine. "In this study, we demonstrated a shared mechanism between the two disorders. Language problems and ASD are complicated and have numerous genetic factors, but we think that many genetic factors related to communication could be the same for specific language impairment and language-impaired autism."

The genetic variations appear to be relevant to both disorders and may indicate a greater level of genetic predisposition for impairments in language ability among individuals with and without ASD in those families.

In an earlier study, the researchers found similarities in language deficit type and severity between language-impaired non-ASD and language-impaired ASD individuals in the same family. The behavioral genetics study, published in Biological Psychiatry, found that the same genes active in specific language impairment appear in ASD, but their effect is amplified in ASD. That finding, coupled with this new research, suggests that the two disorders may be on an etiological continuum.
"If further research confirms a genetic link between language impairment and ASD, then we may be able to find out why some family members only develop language impairment while others develop autism," says Dr. Bartlett. "But most of all, we want to know why there is such a range in communication abilities in children with autism—why so many children have language difficulties when it’s not required for the diagnosis."

Provided by Nationwide Children's Hospital


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