

Researchers find alterations of a single gene associated with intellectual disability, epilepsy and autistic features

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(Medical Xpress) -- Virginia Commonwealth University School of Medicine researchers, working with an international team of colleagues, have identified a gene that may play a role in causing a neurodevelopmental disorder that includes intellectual disability, seizures and autism spectrum disorder.

The molecular findings may one day change the way diagnostic laboratories assess the [DNA](#) of individuals being evaluated for certain neurodevelopmental disorders or autism spectrum disorder, with more careful consideration placed on very small [genetic changes](#) that may affect [gene expression](#).

Chromosomal microdeletions - the loss of an extremely small piece of genetic material that can result in a mutation - have been observed in people with neurodevelopmental disorders or autism spectrum disorders. There has been little research done to more closely evaluate the individual genetic contributors within these regions, until now.

In a study published online in the October issue of the *American Journal of Human Genetics*, the team led by VCU and comprised of researchers from the U.S., Europe, Canada and Australia, and more than 20 institutions, conducted a large-scale characterization of a microdeletion syndrome previously implicated in neurodevelopmental abnormalities. The findings suggest that the impact of both notable and slight mutations

at a select locus may account for approximately 1 percent of the overall disease risk in autism spectrum disorders.

According to lead investigator, Sarah H. Elsea, Ph.D., associate professor in the VCU Departments of Pediatrics and Human and Molecular Genetics, the team identified the gene, MBD5, as the sole causative gene for 2q23.1 deletion syndrome. MBD5 functions in regulating the expression of many genes and is responsible for the core clinical features in these individuals, including intellectual disability, epilepsy and autism spectrum disorder. They have also shown that there is an association between autism and MBD5.

"This project revealed that alterations of a single gene can be sufficient to cause a [neurodevelopmental disorder](#) that includes [intellectual disability](#), [seizures](#) and autism spectrum disorder," Elsea said. "Further study of this gene could help us understand the disease process that can lead to autism and other developmental abnormalities. The findings indicate that approximately 1 percent of autism spectrum disorder cases may be attributed variations in this gene.

"One outcome of these studies is that, depending on the type of mutation, the severity of the disorder may be predicted and interventions started early, which may improve outcomes for all individuals. We hope that these findings will push diagnostic laboratories to change the way they assess [genes](#) in the diagnosis of neurodevelopmental disorders, which will improve diagnosis and inform parents regarding familial risk for autism spectrum disorder."

Sureni Mullegama, a grad student in Elsea's lab and first co-author on the study, will be traveling to present these findings to colleagues during the joint American Society of Human Genetics/International Congress of Human Genetics in Montreal, Canada being held from October 11 to 15.

Elsa's team is continuing to explore the genetic pathways that are affected when MBD5 is not in sufficient quantity or functioning properly to gain further understanding of how neurodevelopmental disorders, including [autism spectrum disorder](#) and other more severe genetic syndromes, are related on a biochemical level.

Provided by Virginia Commonwealth University

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