Genetic mutations called structural variants linked to autism spectrum disorders

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A research team led by ORNL's Michael Garvin, left, and David Kainer discovered genetic mutations called structural variants and linked them to autism spectrum disorders, demonstrating an approach that could be used to develop better diagnostics and drug therapies. Credit: Carlos Jones/ORNL, U.S. Dept. of Energy

An Oak Ridge National Laboratory-led research team discovered genetic

mutations that underlie autism using a new approach that could lead to better diagnostics and drug therapies.

Their study is published in *Human Genetics and Genomics Advances*.

Scientists estimate 80% of autism is inherited, but they have yet to identify causative genes.

"We realized the value of unexplored heritable information from others' research," said ORNL's Michael Garvin. Garvin and colleagues focused on genomic mutations called structural variants and established a direct link to autism traits.

The key was observing that many structural variants are excluded because they often display nontraditional inheritance patterns. By focusing on these variants, ORNL scientists found a mutation in the ACMSD gene that is associated with nonverbal types of autism. They then used artificial intelligence and high-performance computing to find additional variants related to three autism subtypes.

"We've established a workflow for using this often-ignored data that can be applied not only to autism, but also to other disorders," said ORNL's David Kainer.

More information: David Kainer et al, Structural variants identified using non-Mendelian inheritance patterns advance the mechanistic understanding of autism spectrum disorder, *Human Genetics and Genomics Advances* (2022). DOI: 10.1016/j.xhgg.2022.100150

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