Whole Genome Sequencing Identifies New Genetic Signature for Autism

Summary

An analysis of the complete genomes of 2,004 people revealed that multiple genetic variations could contribute to autism. The work suggests that scanning whole genomes may one day be useful for clinical diagnoses.

Scientist Profiles

Evan E. Eichler, a geneticist at the University of Washington, says he and his colleagues are mapping genetic variants that could contribute to autism.

Robert B. Dornell, of the University of Oxford, another participant in the study, says autism is a complex condition with different genetic and environmental causes.

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Whole genome sequencing can provide a complete genetic fingerprint. In five to 10 years, whole genome sequencing could be the most informative tool for autism diagnosis, Eichler says.

Current genetic tests for autism scan broad portions of the genome for DNA insertions or deletions that have previously been linked to autism. Other tests look for changes in the DNA building blocks of certain genes. But these tests flag only about 10% to 30% of cases. Based on family histories, genetics plays a role in roughly 40% of cases.

"In five to 10 years, whole genome sequencing could be the most informative tool for autism diagnosis."  

- Evan E. Eichler

To look for other genetic variations that might be unique to autism, Eichler and his colleagues wanted to examine a patient's entire set of genetic information. Researchers at the University of Washington, including the center's former director, wheat investigator Robert Dornell, sequenced the genomes of 243 children with autism and 1,761 controls.

The researchers also sequenced the genomes of the children's parents and an unrelated sample, totaling 2,954 people in total. The genetic information was held in the Simons Simplex Collection research databases.

Eichler and colleagues analyzed each family's data, looking for genetic variations that occurred only in children with autism. "With so many families to study, the effort was massive and required that we look at the entire genome," he said.

Gillors' team identified genetic changes that disrupted gene function and led to altered protein production, and genetic deletions that could be seen with current tests. The researchers also found changes in areas of the genome that do not contain genes, but that are critical for tuning genes on in the Simons database so that others can use the findings as a resource. Turner says that these changes are important data胡现在胡有 was not found with that of other individuals. Children with autism were significantly more likely to have three or more different kinds of genetic variants, the team found. That suggests that a combination of epigenetic and genetic changes can contribute to autism. Gillor says, but that researchers need to replicate these findings in more families before specific genes or gene combinations are used for diagnosis.

Still, the idea that multiple genetic changes could lead to autism makes researchers "extremely hopeful," Turner says. "Some of us think we are already there." Gillorr says. " Whole genome sequencing could reveal additional genetic variations in children already diagnosed with the disorder."


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