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What will my child's life be like? Newly identified genes may help diagnose autism and disability

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There are now 91 genes with known links to autism and intellectual disabilities. from www.shutterstock.com

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Editor's note: The statistical analysis in the paper this article is based on is being queried by independent researchers. A complete statistical reanalysis and formal response by the study authors is being prepared. We will provide an update once this statement is available.

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We have identified 38 new genes that are strongly associated with autism and intellectual disability. Some of the genes appear to be primarily associated with autism, and others primarily associated with intellectual disability.

We also found evidence reinforcing the importance of 53 previously identified genes in autism and intellectual disability.

Improved knowledge of genes involved in autism and intellectual disorders will allow families to better anticipate the needs of their children and guide early learning and social supports. It may also lead to better understanding of the fundamental processes of learning, memory and behaviour.

New genes linked with autism and disability

Unravelling the genetic contribution to autism and intellectual disability is not about blaming parents or categorising children. Rather, it can improve opportunities for accurate diagnosis and early interventions. More than 50% of individuals with autism or intellectual disability do not currently have a precise diagnosis.

We assessed DNA from more than 11,700 patients from 15 centres across seven countries and four continents. The patients recruited for this study had a clinical diagnosis of autism and/or intellectual disability. We also assessed DNA from 2,800 individuals without disability, our control group.

Of the 38 new genes identified, mutations in eight in particular were most strongly linked to patients with autism, often high-functioning autism (characterised by relatively high intellectual ability). Mutations in 17 of the new genes were most strongly linked with intellectual disability.

For the majority of the genes, however, it was not possible to assign them as being more commonly associated with either autism or intellectual disability.

We were surprised that the majority of the mutations – about 65% – were inherited from one or the other parent, who were unaffected or much less severely affected with autism or intellectual disability. These mutations do not appear to be sufficient to cause autism or intellectual disability on their own, interacting with other genetic or non-genetic factors to do so.

For now, we do not know which other factors partner with genetic mutations to cause disability, or whether factors may protect the unaffected individuals with these mutations from having autism or a disability. Searching for these “safeguards” or “accomplices” is important, as their identification may help us to develop better diagnostic and therapeutic tools for these conditions in the future.

Genetic information can help family decisions

This study provides strong evidence for the role of 91 genes in autism and intellectual disabilities. That opens up the possibility of using DNA-sequencing technologies to search for these genes in individuals with undiagnosed disabilities across the world.

We can also plan new approaches to correct the functioning of these genes or minimise consequences of their malfunction.

Precise genetic knowledge relating to autism or intellectual disability is expected to help families and health care providers, allowing them to better appreciate the extent of clinical complications typically associated with a specific gene mutation. Such information is very important, especially when a DNA

diagnosis of a developmental disability is reached at an early age and many questions about future development of the child are asked and need to be managed.

It could also lead to a precise test for families looking for guidance in future reproductive decisions.

Genes and environment shape brain function

A high proportion of autism and intellectual disability still remains undiagnosed. This current study adds to existing evidence that a proportion of these conditions do have a genetic component. More genes will undoubtedly be discovered in the future.

However we also hope to better understand not just the genetic contribution but also the role of the environment in shaping how autism and intellectual disability develop.

Better knowledge of the genetic and non-genetic factors that shape brain development will help clinical management, education and support for individuals and families.

The majority of this work was directed and performed by Evan Eichler.