

NEWS

Genetics: Language gene has limited role in autism

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Protein loss: Levels of CNTNAP2, which has been linked to autism in some genetic studies, are lower in brains from some people with autism than in controls.

Variants in a language gene linked to autism are only modestly associated with the disorder in a large sample, according to a study published 17 October in *PLoS One*¹.

CNTNAP2 codes for a protein involved in cell-to-cell communication that binds to **FOXP2**, a language gene **also linked to autism**. Common variants in CNTNAP2 have been implicated in **autism, language delay** and deficits in the **connections between brain regions**.

Mice lacking CNTNAP2 have **autism-like features**, including **repetitive behaviors** and an imbalance between activating and dampening signals in the brain.

In the new study, researchers aimed to strengthen these associations by looking at more than 2,000 variants in CNTNAP2 in more than 500 families with a history of autism: 186 of these families are multiplex, meaning they have more than one child with autism, and the other 323 are simplex, having only one child with the disorder. Individuals with autism from simplex families tend to carry either spontaneous mutations or common mutations with small effects.

The researchers found 2 of these 2,000 variants to be associated with autism — one in the multiplex and one in the simplex families. The association disappeared after multiple rounds of statistical

testing, however.

When the researchers combined the data from both sets of families, they were able to identify two new variants in CNTNAP2 as associated with autism. They then looked at three of these four variants, along with three others previously linked to autism, in an independent sample of 572 multiplex and 1,479 simplex families. They were not able to replicate the associations in this sample.

To further investigate CNTNAP2's role in autism, the researchers looked at postmortem brain tissue from 39 people who had autism and 44 controls. They found that the brains from people with autism are more likely to have low expression of CNTNAP2 than those from controls.

References:

1: Sampath S. *et al.* *PLoS One* **8**, e77906 (2013) [PubMed](#)