

TOOLBOX

Analysis probes literature for autism-linked gene culprits

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By combining bioinformatics techniques with an analysis of gene expression, researchers have identified 30 candidate genes for autism, according to a study published 28 May in *Translational Psychiatry*¹.

These genes include five that previous studies have linked to autism, including **CNTN4**, which helps organize **synapses**, the junctions between neurons. Many of the other genes are linked to related neurological disorders such as schizophrenia or intellectual disability.

To find genes associated with autism, researchers often look to see whether certain genetic markers are more common in people with autism than in controls. These markers may range from alterations in single DNA nucleotides to duplications or deletions of regions of a chromosome, called **copy number variations** (CNVs).

If people with autism are more likely than controls to inherit these markers, they are also more likely to inherit nearby genes. And it is possible that the linked genes also contribute to autism risk.

To explore this possibility, the researchers pinpointed 67 genetic regions that have a strong statistical link to autism in the published literature. They then identified all the genetic variants present in the ten million nucleotides before or after these regions.

Using data from the **Autism Genetic Resource Exchange** — a database of families that have more than one child with autism — the researchers built a genetic map. This map organizes genes based on the likelihood of being inherited together when chromosomes separate during reproduction. Because autism is four times more common in men than in women, the researchers looked specifically at male genomes.

Their analysis identified 1,426 genes that are linked with the 67 autism markers. The researchers then looked to see which of these genes are expressed at higher or lower levels in people with autism than in controls, in both blood and postmortem brain samples. This approach narrowed

down the list to 30 candidate genes.

The fact that five of these are already strongly linked to autism provides support for the method, the researchers say. The other 25 may also turn out to be important for the disorder, they suggest.

References:

1: Vardarajan B.N. *et al. Transl. Psychiatry* **3**, e262 (2013) **PubMed**