NEWS

Genetics: Middle East study tags intellectual disability genes

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By focusing on recessive mutations inherited from both parents, researchers have identified 50 new candidate genes for intellectual disability, according to a study published 21 September in *Nature*[1].

The results support research on the role of recessive mutations — those that need to be present on both copies of a gene to have an effect — in intellectual disability and **autism**.

For example, researchers originally identified mutations in **CNTNAP2**, an important autism candidate gene, in an Amish population that has a higher-than-normal rate of recessive mutations[2].

In the new study, researchers sequenced the genomes of 136 families in Iran with a history of

intellectual disability. Because **marriage between first cousins** is common among families in the Middle East, recessive mutations occur more frequently in these families than in the west.

To identify recessive mutations, the researchers sequenced the **exomes**, or protein-coding regions of the genome, in chromosomal regions that are likely to carry recessive mutations.

Of the 136 families, 115 harbor recessive mutations that are a likely cause of their intellectual disability, the study found. Of these, 78 families have a mutation in a single gene, and 23 of the genes are known to contribute to intellectual disability.

The researchers also identified 50 new candidate genes for intellectual disability, including **CACNA1G**, a calcium channel gene implicated in autism and the related disorder **Timothy syndrome**.

The candidate genes belong to pathways involved in gene expression, the cell cycle and signaling between neurons.

References:

1: Najmabadi H. *et al. Nature* **478**, 57-63 (2011) **PubMed**

2: Strauss K.A. *et al. N. Engl. J. Med.* **354**, 1370-1377 (2006) **PubMed**