CTNND2 gene

catenin delta 2

Normal Function

The *CTNND2* gene provides instructions for making a protein called delta-catenin. This protein is active in the nervous system, where it likely helps cells stick together (cell adhesion) and plays a role in cell movement. In the developing brain, it may help guide nerve cells to their proper positions as part of a process known as neuronal migration.

In mature nerve cells, delta-catenin is located in specialized outgrowths called dendrites. Dendrites branch out from the cell and receive information from nearby nerve cells. This information is relayed across synapses, which are junctions between nerve cells where cell-to-cell communication occurs. Delta-catenin appears to play a crucial role in the function of synapses.

Health Conditions Related to Genetic Changes

Autism spectrum disorder

MedlinePlus Genetics provides information about Autism spectrum disorder

Cri-du-chat syndrome

The *CTNND2* gene is located in a region of chromosome 5 that is often deleted in people with cri-du-chat syndrome. As a result of this deletion, many people with this condition are missing one copy of the *CTNND2* gene in each cell. The loss of this gene may cause severe intellectual disability in some affected individuals. Researchers suspect that intellectual disability could result from a disruption of neuronal migration during the early development of the nervous system.

People with cri-du-chat syndrome who do not have a deletion of the *CTNND2* gene tend to have milder intellectual disability or normal intelligence.

Other Names for This Gene

- catenin (cadherin-associated protein), delta 2
- CTND2_HUMAN
- GT24

- neural plakophilin-related armadillo-repeat protein
- neurojungin
- NPRAP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CTNND2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1501[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CTNND2%5BTIAB%5D%29+OR+%28%28neural+plakophilin-related+armadillo-repeat+protein%5BTIAB%5D%29+OR+%28NPRAP%5BTIAB%5D%29+OR+%28neurojungin%5BTIAB%5D%29+OR+%28delta-catenin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CATENIN, DELTA-2; CTNND2 (https://omim.org/entry/604275)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1501)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CTNND2[gene])

References

- Israely I, Costa RM, Xie CW, Silva AJ, Kosik KS, Liu X. Deletion of theneuron-specific protein delta-catenin leads to severe cognitive and synapticdysfunction. Curr Biol. 2004 Sep 21;14(18):1657-63. doi:10.1016/j.cub.2004.08.065. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15380068)
- Kosik KS, Donahue CP, Israely I, Liu X, Ochiishi T. Delta-catenin at thesynaptic-adherens junction. Trends Cell Biol. 2005 Mar;15(3):172-8. doi:10.1016/j.tcb.2005.01.004. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15752981)
- Medina M, Marinescu RC, Overhauser J, Kosik KS. Hemizygosity of delta-catenin(CTNND2) is associated with severe mental retardation in cri-du-chat syndrome. Genomics. 2000 Jan 15;63(2):157-64. doi: 10.1006/geno.1999.6090. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10673328)

Genomic Location

The *CTNND2* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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