

UNC80 gene

unc-80 homolog, NALCN channel complex subunit

Normal Function

The *UNC80* gene provides instructions for making a large protein that is important in the functioning of a sodium channel called NALCN. Sodium channels transport positively charged sodium atoms (sodium ions) into cells and play a key role in a cell's ability to generate and transmit electrical signals. The UNC80 protein forms a bridge between NALCN and another protein called UNC79; along with several other molecules, these proteins group together to form a functional NALCN channel complex (channelosome). UNC80 also helps locate and stabilize the NALCN channelosome in the cell membrane of nerve cells (neurons). The channelosome helps regulate the activity level (excitability) of these cells.

Health Conditions Related to Genetic Changes

UNC80 deficiency

At least 10 *UNC80* gene mutations have been identified in people with UNC80 deficiency, a disorder that causes severe nervous system and developmental problems that are apparent from birth or early infancy. The mutations that occur in people with this disorder result in absence of the UNC80 protein or production of an abnormal protein. Absence of functional UNC80 protein impairs the stability and function of the NALCN channelosome. Neuron excitability is thought to be improperly regulated as a result, but it is unclear how these changes cause the specific features of UNC80 deficiency.

Other Names for This Gene

- C2orf21
- FLJ33496
- KIAA1843
- protein unc-80 homolog isoform 1
- protein unc-80 homolog isoform 2
- UNC-80
- unc-80 homolog, NALCN activator

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of UNC80 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=285175\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=285175[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28UNC80%5BTIAB%5D%29+OR+%28unc-80+homolog,+NALCN+channel+complex+subunit%5BTIAB%5D%29%29+OR+%28%28UNC-80%5BTIAB%5D%29+OR+%28protein+unc-80+homolog+isoform+1%5BTIAB%5D%29+OR+%28protein+unc-80+homolog+isoform+2%5BTIAB%5D%29+OR+%28unc-80+homolog,+NALCN+activator%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

- UNC80 HOMOLOG, NALCN CHANNEL COMPLEX SUBUNIT; UNC80 (<https://omim.org/entry/612636>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/285175>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=UNC80\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=UNC80[gene]))

References

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Genomic Location

The *UNC80* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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