



## CHN1 gene

chimerin 1

### Normal Function

The *CHN1* gene provides instructions for making two very similar proteins called  $\alpha$ 1-chimaerin and  $\alpha$ 2-chimaerin. These proteins play an important role in the early development of the nervous system. In particular, they help regulate complex chemical signaling pathways during the formation and development of nerve cells (neurons). These proteins help guide the growth of axons and dendrites, which are specialized extensions of neurons that transmit and receive nerve impulses throughout the nervous system.

The CHN1 proteins, particularly  $\alpha$ 2-chimaerin, appear to be critical for the formation of certain nerves in the head and face. Specifically, they are necessary for the development and function of cranial nerve VI and, to a lesser extent, cranial nerve III. These nerves emerge from the brain and control several of the muscles that surround the eyes (extraocular muscles). The extraocular muscles direct eye movement and determine the position of the eyes.

### Health Conditions Related to Genetic Changes

#### Isolated Duane retraction syndrome

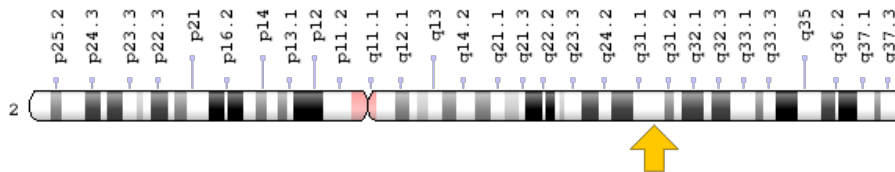
At least seven mutations in the *CHN1* gene have been identified in families with isolated Duane retraction syndrome. Researchers believe that the features of this condition result from changes in one of the two versions of the CHN1 protein,  $\alpha$ 2-chimaerin. Each identified mutation changes a single protein building block (amino acid) in  $\alpha$ 2-chimaerin. Although several mutations also affect  $\alpha$ 1-chimaerin, researchers think that changes in  $\alpha$ 1-chimaerin alone do not cause isolated Duane retraction syndrome.

*CHN1* mutations alter the structure and function of  $\alpha$ 2-chimaerin in the developing nervous system. These genetic changes appear to overactivate the protein, which disrupts the normal growth of neurons in certain parts of the brain. As a result, cranial nerves VI and III and the extraocular muscles they control do not develop normally. Abnormal development and function of these muscles leads to the characteristic features of isolated Duane retraction syndrome, including restricted eye movement and related problems with vision.

## Chromosomal Location

Cytogenetic Location: 2q31.1, which is the long (q) arm of chromosome 2 at position 31.1

Molecular Location: base pairs 174,798,809 to 175,005,381 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- A-chimaerin
- alpha-chimerin
- ARHGAP2
- Chimerin 1 (GTPase-activating protein, rho, 2)
- CHIN\_HUMAN
- CHN
- DURS2
- N-chimaerin
- n-chimerin
- Rho GTPase-activating protein 2
- RHOGAP2

## Additional Information & Resources

### Clinical Information from GeneReviews

- Duane Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1190>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CHN1%5BTIAB%5D%29+OR+%28chimerin+1%5BTIAB%5D%29%29+OR+%28%28alpha-chimerin%5BTIAB%5D%29+OR+%28ARHGAP2%5BTIAB%5D%29+OR+%28DURS2%5BTIAB%5D%29+OR+%28n-chimerin%5BTIAB%5D%29+OR+%28N-chimaerin%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+1800+days%22%5Bdp%5D>

## Catalog of Genes and Diseases from OMIM

- CHIMERIN 1  
<http://omim.org/entry/118423>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_CHN1.html](http://atlasgeneticsoncology.org/Genes/GC_CHN1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=CHN1%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:1943](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:1943)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:1123>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1123>
- UniProt  
<https://www.uniprot.org/uniprot/P15882>

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