

## CHN1 gene

chimerin 1

### Normal Function

The *CHN1* gene provides instructions for making a protein called N-chimaerin. This protein plays an important role in the early development of the nervous system. In particular, it helps regulate complex signaling pathways during the development of nerve cells (neurons). N-chimaerin helps guide the growth of axons and dendrites, which are specialized extensions of neurons that transmit and receive nerve impulses throughout the nervous system.

N-chimaerin appears to be critical for the formation of certain nerves in the head and face, known as cranial nerves. The protein is necessary for the development and function of cranial nerve VI and, to a lesser extent, cranial nerve III. These two 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

Variants (also called mutations) in the *CHN1* gene have been identified in families with isolated Duane retraction syndrome. This condition prevents outward movement of the eye (toward the ear), and in some cases it may also limit inward eye movement (toward the nose). This condition is referred to as "isolated" because it causes eye problems with no other signs or symptoms.

*CHN1* gene variants alter the structure and function of N-chimaerin in the developing nervous system. These variants are called "gain of function" variants because they appear to enhance the activity of the protein. This can disrupt the normal growth of neurons in certain parts of the brain. As a result, cranial nerves VI and III do not develop normally and cannot effectively control the extraocular muscles. Impaired function of these muscles leads to the characteristic features of isolated Duane retraction syndrome.

## Other Names for This Gene

- A-chimaerin
- alpha-chimerin
- CHIN\_HUMAN
- CHN
- N-chimaerin
- n-chimerin
- RHOGAP2

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of CHN1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1123\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1123[geneid]))

### Scientific Articles on PubMed

- PubMed ([https://pubmed.ncbi.nlm.nih.gov/?term=\(\(CHN1%5BTIAB%5D\)+OR+\(chimerin+1%5BTIAB%5D\)+OR+\(n-chimerin%5BTIAB%5D\)+OR+\(N-chimaerin%5BTIAB%5D\)\)+AND+\(\(Genes%5BMH%5D\)+OR+\(Genetic+Phenomena%5BMH%5D\)\)+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+5000+days%22%5Bdp%5D\)](https://pubmed.ncbi.nlm.nih.gov/?term=((CHN1%5BTIAB%5D)+OR+(chimerin+1%5BTIAB%5D)+OR+(n-chimerin%5BTIAB%5D)+OR+(N-chimaerin%5BTIAB%5D))+AND+((Genes%5BMH%5D)+OR+(Genetic+Phenomena%5BMH%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+5000+days%22%5Bdp%5D)))

### Catalog of Genes and Diseases from OMIM

- CHIMERIN 1; CHN1 (<https://omim.org/entry/118423>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1123>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CHN1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CHN1[gene]))

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## Genomic Location

The *CHN1* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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