

FRMD7 gene

FERM domain containing 7

Normal Function

The *FRMD7* gene provides instructions for making a protein whose exact function is unknown. This protein is found in many tissues, but it is most abundant in areas of the brain that control eye movement (such as the midbrain and cerebellum) and in the light-sensitive tissue at the back of the eye (retina). The FRMD7 protein likely plays a role in the development of nerve cells in these areas of the brain and the retina.

Health Conditions Related to Genetic Changes

X-linked infantile nystagmus

More than 35 mutations in the *FRMD7* gene have been found to cause X-linked infantile nystagmus. Most of these mutations change single protein building blocks (amino acids) in the FRMD7 protein. Mutations in the *FRMD7* gene likely lead to the production of a protein that is unstable and that cannot perform its normal function. A lack of functional FRMD7 protein is thought to disrupt the development of nerve cells in the retina and areas of the brain that control eye movement. Abnormal development of these nerve cells likely causes the involuntary side-to-side eye movements that are characteristic of X-linked infantile nystagmus.

Other Names for This Gene

- FRMD7_HUMAN
- NYS1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FRMD7 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=90167\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=90167[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28FRMD7%5BTIAB%5D%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

- FERM DOMAIN-CONTAINING PROTEIN 7; FRMD7 (<https://omim.org/entry/300628>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/90167>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FRMD7\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FRMD7[gene]))

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Genomic Location

The *FRMD7* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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