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.

Chromosomal Location
Cytogenetic Location: Xq26.2, which is the long (q) arm of the X chromosome at position 26.2
Molecular Location: base pairs 132,074,926 to 132,128,022 on the X chromosome (Homo sapiens Updated Annotation Release 109.20191205, GRCh38.p13) (NCBI)
Other Names for This Gene

• FRMD7_HUMAN
• NYS1

Additional Information & Resources

Clinical Information from GeneReviews

• FRMD7-Related Infantile Nystagmus
  https://www.ncbi.nlm.nih.gov/books/NBK3822

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28FRMD7%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%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
  http://omim.org/entry/300628

Research Resources

• Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FRMD7.html
• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FRMD7%5Bgene%5D
• HGNC Gene Symbol Report
• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:90167
• NCBI Gene
• UniProt
  https://www.uniprot.org/uniprot/Q6ZUT3
Sources for This Summary

- OMIM: FERM DOMAIN-CONTAINING PROTEIN 7
  http://omim.org/entry/300628

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19072571

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18431453
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2324116/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17846367

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17013395
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2592600/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17893669

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17768376

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