



X-linked infantile nystagmus

X-linked infantile nystagmus is a condition characterized by abnormal eye movements. Nystagmus is a term that refers to involuntary side-to-side movements of the eyes. In people with this condition, nystagmus is present at birth or develops within the first six months of life. The abnormal eye movements may worsen when an affected person is feeling anxious or tries to stare directly at an object. The severity of nystagmus varies, even among affected individuals within the same family. Sometimes, affected individuals will turn or tilt their head to compensate for the irregular eye movements.

Frequency

The incidence of all forms of infantile nystagmus is estimated to be 1 in 5,000 newborns; however, the precise incidence of X-linked infantile nystagmus is unknown.

Causes

Mutations in the *FRMD7* gene cause X-linked infantile nystagmus. The *FRMD7* gene provides instructions for making a protein whose exact function is unknown. This protein is found mostly in areas of the brain that control eye movement and in the light-sensitive tissue at the back of the eye (retina). Research suggests that *FRMD7* gene mutations cause nystagmus by disrupting the development of certain nerve cells in the brain and retina.

In some people with X-linked infantile nystagmus, no mutation in the *FRMD7* gene has been found. The genetic cause of the disorder is unknown in these individuals. Researchers believe that mutations in at least one other gene, which has not been identified, can cause this disorder.

Inheritance Pattern

This condition is inherited in an X-linked pattern. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome, one of the two sex chromosomes in each cell. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two copies of the X chromosome), one altered copy of the gene in each cell can cause the condition, although affected females may experience less severe symptoms than affected males. Approximately half of the females with only one altered copy of the *FRMD7* gene in each cell have no symptoms of this condition.

Other Names for This Condition

- congenital motor nystagmus
- *FRMD7*-related infantile nystagmus

- idiopathic infantile nystagmus
- NYS1
- X-linked congenital nystagmus
- X-linked idiopathic infantile nystagmus

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Infantile nystagmus, X-linked
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1839580/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22X-linked+congenital+nystagmus%22+OR+%22Nystagmus%2C+Congenital%22+OR+%22Congenital+Nystagmus%22>

Other Diagnosis and Management Resources

- GeneReview: FRMD7-Related Infantile Nystagmus
<https://www.ncbi.nlm.nih.gov/books/NBK3822>
- MedlinePlus Encyclopedia: Nystagmus
<https://medlineplus.gov/ency/article/003037.htm>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Nystagmus
<https://medlineplus.gov/ency/article/003037.htm>
- Health Topic: Eye Movement Disorders
<https://medlineplus.gov/eyemovementdisorders.html>

Genetic and Rare Diseases Information Center

- Nystagmus 1, congenital, X- linked
<https://rarediseases.info.nih.gov/diseases/2969/nystagmus-1-congenital-x--linked>

Educational Resources

- American Optometric Association: Nystagmus
<https://www.aoa.org/404?aspxerrorpath=%2fpatients-and-public%2fgood-vision-throughout-life%2fchildrens-vision%2finfant-vision-birth-to-24-months-of-age>
- MalaCards: x-linked infantile nystagmus
https://www.malacards.org/card/x_linked_infantile_nystagmus
- Orphanet: Congenital idiopathic nystagmus
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=651

Patient Support and Advocacy Resources

- Royal National Institute of Blind People (UK): Nystagmus
<https://www.rnib.org.uk/eye-health/eye-conditions/nystagmus>
- University of Kansas Medical Center Resource List: Visual impairment
<http://www.kumc.edu/gec/support/visual.html>

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=%28%28x-linked+congenital+nystagmus%5BTIAB%5D%29+OR+%28congenital+motor+nystagmus%5BTIAB%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

- NYSTAGMUS 1, CONGENITAL, X-LINKED
<http://omim.org/entry/310700>

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