X-linked infantile nystagmus

Description

X-linked infantile nystagmus is a condition characterized by abnormal eye movements. Nystagmus is a term that refers to involuntary side-to-side, up-and-down, or circular movements of the eyes. In people with X-linked infantile nystagmus, the movements are typically side-to-side. In individuals 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. Some affected individuals will experience involuntary changes in the direction of their eye movements (periodic alternating nystagmus). 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. Individuals with X-linked infantile nystagmus may have other eye abnormalities. For example, the region at the back of the eye responsible for sharp central vision may be underdeveloped (foveal hypoplasia).

Frequency

The incidence of X-linked infantile nystagmus is estimated to be 4.4 in 10,000 individuals.

Causes

Variants (also called 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 variants cause nystagmus by disrupting the development of certain nerve cells in the brain and retina.

In some people with X-linked infantile nystagmus, no variant in the *FRMD7* gene has been found. The genetic cause of the disorder is unknown in these individuals. Researchers believe that variants in at least one other gene, which has not been identified, can cause this disorder. Nystagmus can also occur in people with other X-linked eye conditions, such as X-linked ocular albinism. Such cases are caused by changes in genes associated with the particular condition.

Learn more about the gene associated with X-linked infantile nystagmus

- FRMD7

Inheritance

X-linked infantile nystagmus is inherited in an X-linked pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In individuals with one X chromosome (typical for males), a variant in the only copy of the gene in each cell is sufficient to cause the condition. In people with two copies of the X chromosome (typical for females), one altered copy of the gene can cause the condition, although the features may be less severe than in individuals with two altered copies. Some of these individuals may have no signs or symptoms at all; approximately half of females with only one altered copy of the *FRMD7* gene 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

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Nystagmus 1, congenital, X-linked (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1839580/)

Genetic and Rare Diseases Information Center

- Nystagmus 1, congenital, X- linked (https://rarediseases.info.nih.gov/diseases/2969/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22X-linked infantile nystagmus%22)

Catalog of Genes and Diseases from OMIM

- NYSTAGMUS 1, CONGENITAL, X-LINKED; NYS1 (https://omim.org/entry/310700)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=((x-linked+congenital+nystagmus%5BTIAB%5D)+OR+(FRMD7+nystagmus%5BTIAB%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+6000+days%22%5Bdp%5D)

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