X-linked congenital stationary night blindness

Description

X-linked congenital stationary night blindness is a disorder of the retina, which is a specialized tissue at the back of the eye that detects light and color. People with this condition typically have difficulty seeing in low light (night blindness). They also have other vision problems, including increased sensitivity to light (photophobia), loss of sharpness (reduced visual acuity), severe nearsightedness (high myopia), involuntary movements of the eyes (nystagmus), and eyes that do not look in the same direction (strabismus). Color vision is typically not affected in people with X-linked congenital stationary night blindness.

The vision problems associated with X-linked congenital stationary night blindness are congenital, which means they are present from birth. The vision problems also tend to remain stable (stationary) over time.

Researchers have identified two major types of X-linked congenital stationary night blindness: the complete form and the incomplete form. The types have very similar signs and symptoms. However, everyone with the complete form has night blindness, while not all people with the incomplete form have night blindness. The types are distinguished by their genetic causes and by the results of a test called an electroretinogram, which measures the function of the retina.

Frequency

The prevalence of X-linked congenital stationary night blindness is unknown. The incomplete form is more common than the complete form.

Causes

Variants (also called mutations) in the *NYX* gene cause the complete form of X-linked congenital stationary night blindness, and variants in the *CACNA1F* gene cause the incomplete form. The proteins produced from these genes play critical roles in the retina.

Within the retina, the CACNA1F protein is located on light-detecting cells called photoreceptors. The NYX protein is located on cells called bipolar cells, which relay signals to other retinal cells. The retina contains two types of photoreceptor cells: rods and cones. Rods are needed for vision in low light. Cones are needed for vision in bright

light, including color vision. The NYX and CACNA1F proteins ensure that visual signals are passed from rods and cones to bipolar cells, which is an essential step in the transmission of visual information from the eyes to the brain.

Variants in the *NYX* or *CACNA1F* gene disrupt the transmission of visual signals between photoreceptors and bipolar cells, which impairs vision. In people with the complete form of X-linked congenital stationary night blindness, the rod pathway is severely disrupted, while the cone pathway is only mildly affected. In people with the incomplete form of the condition, the rod and cone pathways are both affected, although the affected person does have the ability to detect some light.

A particular variant in the *CACNA1F* gene has been found to cause X-linked congenital stationary night blindness in individuals who are of Dutch-German Mennonite descent. Similarly, certain *NYX* gene variants appear to be a frequent cause of X-linked congenital stationary night blindness in people who live in the United States and in people who live in Belgium and are of Flemish descent.

Learn more about the genes associated with X-linked congenital stationary night blindness

- CACNA1F
- NYX

Inheritance

X-linked congenital stationary night blindness is inherited in an X-linked recessive pattern. The *NYX* and *CACNA1F* genes are located on the X chromosome, which is one of the two sex chromosomes. 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 X chromosomes), a variant would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that both copies of the gene would be altered, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

In X-linked recessive inheritance, a female with one altered copy of the gene on one of the two X chromosomes is called a carrier. Carriers of an *NYX* or *CACNA1F* gene variant can pass on the altered gene, but most do not develop any of the vision problems associated with X-linked congenital stationary night blindness. However, carriers may have retinal changes that can be detected with an electroretinogram.

Other Names for This Condition

- X-linked CSNB
- XLCSNB

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Congenital stationary night blindness (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0339535/)
- Genetic Testing Registry: Congenital stationary night blindness 1A (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3495587/)
- Genetic Testing Registry: Congenital stationary night blindness 2A (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1848172/)

Genetic and Rare Diseases Information Center

- Congenital stationary night blindness (https://rarediseases.info.nih.gov/diseases/3995/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 congenital stationary night blindness%22)

Catalog of Genes and Diseases from OMIM

- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A; CSNB1A (https://omim.org/entry/310500)
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 2A; CSNB2A (https://omim.org/entry/300071)

Scientific Articles on PubMed

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

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