X-linked congenital stationary night blindness

Description

X-linked congenital stationary night blindness is a disorder of the retina, which is the 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 loss of sharpness (reduced 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 by this disorder.

The vision problems associated with this condition are congenital, which means they are present from birth. They 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 cause and by the results of a test called an electroretinogram, which measures the function of the retina.

Frequency

The prevalence of this condition is unknown. It appears to be more common in people of Dutch-German Mennonite descent. However, this disorder has been reported in families with many different ethnic backgrounds. The incomplete form is more common than the complete form.

Causes

Mutations in the *NYX* and *CACNA1F* genes cause the complete and incomplete forms of X-linked congenital stationary night blindness, respectively. The proteins produced from these genes play critical roles in the retina.

Within the retina, the NYX and CACNA1F proteins are located on the surface of light-detecting cells called photoreceptors. 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 other retinal cells called bipolar cells,

which is an essential step in the transmission of visual information from the eyes to the brain.

Mutations in the *NYX* or *CACNA1F* gene disrupt the transmission of visual signals between photoreceptors and retinal bipolar cells, which impairs vision. In people with the complete form of X-linked congenital stationary night blindness (resulting from *NYX* mutations), the function of rods is severely disrupted, while the function of cones is only mildly affected. In people with the incomplete form of the condition (resulting from *CACNA1F* mutations), rods and cones are both affected, although they retain some ability to detect light.

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

- CACNA1F
- NYX

Inheritance

This condition 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 mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, 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 in each cell is called a carrier. Carriers of an *NYX* or *CACNA1F* mutation can pass on the mutated 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=%28%28x-linked+congenital+stationary+night+blindness%5BTIAB%5D%29+OR+%28x-linked+csnb%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

References

- Allen LE, Zito I, Bradshaw K, Patel RJ, Bird AC, Fitzke F, Yates JR, Trump D, Hardcastle AJ, Moore AT. Genotype-phenotype correlation in British families withX linked congenital stationary night blindness. Br J Ophthalmol. 2003Nov;87(11):1413-20. doi: 10.1136/bjo.87.11.1413. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14609846) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1771890/)
- Bech-Hansen NT, Naylor MJ, Maybaum TA, Pearce WG, Koop B, Fishman GA,

Mets M,Musarella MA, Boycott KM. Loss-of-function mutations in a calcium-channelalpha1-subunit gene in Xp11.23 cause incomplete X-linked congenital stationarynight blindness. Nat Genet. 1998 Jul;19(3):264-7. doi: 10.1038/947. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9662400)

- Bech-Hansen NT, Naylor MJ, Maybaum TA, Sparkes RL, Koop B, Birch DG, BergenAA, Prinsen CF, Polomeno RC, Gal A, Drack AV, Musarella MA, Jacobson SG, YoungRS, Weleber RG. Mutations in NYX, encoding the leucine-rich proteoglycannyctalopin, cause X-linked complete congenital stationary night blindness. NatGenet. 2000 Nov;26(3):319-23. doi: 10.1038/81619. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11062471)
- Boycott KM, Pearce WG, Bech-Hansen NT. Clinical variability among patientswith incomplete X-linked congenital stationary night blindness and a foundermutation in CACNA1F. Can J Ophthalmol. 2000 Jun;35(4):204-13. doi:10.1016/s0008-4182(00)80031-9. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10900517)
- Jacobi FK, Andreasson S, Langrova H, Meindl A, Zrenner E, Apfelstedt-Sylla E, Pusch CM. Phenotypic expression of the complete type of X-linked congenitalstationary night blindness in patients with different mutations in the NYX gene.Graefes Arch Clin Exp Ophthalmol. 2002 Oct;240(10):822-8. doi:10.1007/s00417-002-0562-z. Epub 2002 Sep 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12397430)
- MacDonald IM, Hoang S, Tuupanen S. X-Linked Congenital Stationary NightBlindness. 2008 Jan 16 [updated 2019 Jul 3]. In: Adam MP, Feldman J, Mirzaa GM,Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R) [Internet]. Seattle (WA): University of Washington,Seattle; 1993-2024. Available from http://www.ncbi.nlm.nih.gov/books/NBK1245/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301423)
- Pusch CM, Zeitz C, Brandau O, Pesch K, Achatz H, Feil S, Scharfe C, Maurer J, Jacobi FK, Pinckers A, Andreasson S, Hardcastle A, Wissinger B, Berger W, MeindlA. The complete form of X-linked congenital stationary night blindness is causedby mutations in a gene encoding a leucine-rich repeat protein. Nat Genet. 2000Nov;26(3):324-7. doi: 10.1038/81627. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11062472)
- Strom TM, Nyakatura G, Apfelstedt-Sylla E, Hellebrand H, Lorenz B, Weber BH, Wutz K, Gutwillinger N, Ruther K, Drescher B, Sauer C, Zrenner E, Meitinger T, Rosenthal A, Meindl A. An L-type calcium-channel gene mutated in incompleteX-linked congenital stationary night blindness. Nat Genet. 1998 Jul;19(3):260-3.doi: 10.1038/940. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9662399)

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