X-linked congenital stationary night blindness

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 \textit{CACNA1F} mutations), rods and cones are both affected, although they retain some ability to detect light.

\subsection*{Inheritance Pattern}

This condition is inherited in an X-linked recessive pattern. The \textit{NYX} and \textit{CACNA1F} genes are located on the \textit{X} chromosome, which is one of the two sex chromosomes. In males (who have only one \textit{X} chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two \textit{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 \textit{NYX} or \textit{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.

\subsection*{Other Names for This Condition}

- X-linked CSNB
- XLCSNB

\subsection*{Diagnosis & Management}

\subsubsection*{Genetic Testing Information}

- What is genetic testing? \(\text{primer/testing/genetictesting}\)
- Genetic Testing Registry: Congenital stationary night blindness, type 1A https://www.ncbi.nlm.nih.gov/gtr/conditions/C3495587/

\subsubsection*{Research Studies from ClinicalTrials.gov}

- ClinicalTrials.gov https://clinicaltrials.gov/ct2/results?cond=%22X-linked+congenital+stationary+night+blindness%22
Other Diagnosis and Management Resources

- American Optometric Association: Infant Vision
- GeneReview: X-Linked Congenital Stationary Night Blindness
  https://www.ncbi.nlm.nih.gov/books/NBK1245
- MedlinePlus Encyclopedia: Electroretinography
  https://medlineplus.gov/ency/article/003388.htm
- MedlinePlus Encyclopedia: Eye movements - Uncontrollable
  https://medlineplus.gov/ency/article/003037.htm
- MedlinePlus Encyclopedia: Nearsightedness
  https://medlineplus.gov/ency/article/001023.htm
- MedlinePlus Encyclopedia: Strabismus
  https://medlineplus.gov/ency/article/001004.htm
- MedlinePlus Encyclopedia: Vision - Night Blindness
  https://medlineplus.gov/ency/article/003039.htm

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Electroretinography
  https://medlineplus.gov/ency/article/003388.htm
- Encyclopedia: Eye movements - Uncontrollable
  https://medlineplus.gov/ency/article/003037.htm
- Encyclopedia: Nearsightedness
  https://medlineplus.gov/ency/article/001023.htm
- Encyclopedia: Strabismus
  https://medlineplus.gov/ency/article/001004.htm
- Encyclopedia: Vision - Night Blindness
  https://medlineplus.gov/ency/article/003039.htm
- Health Topic: Vision Impairment and Blindness
  https://medlineplus.gov/visionimpairmentandblindness.html

Genetic and Rare Diseases Information Center

- X-linked congenital stationary night blindness
  https://rarediseases.info.nih.gov/diseases/3995/x-linked-congenital-stationary-night-blindness
Additional NIH Resources

• National Eye Institute: Low Vision
  https://nei.nih.gov/health/lowvision/

Educational Resources

• MalaCards: x-linked congenital stationary night blindness
  https://www.malacards.org/card/x_linked_congenital_stationary_night_blindness

• Orphanet: Congenital stationary night blindness
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=215

Patient Support and Advocacy Resources

• American Foundation for the Blind
  https://www.afb.org/

• Foundation Fighting Blindness
  https://www.fightingblindness.org/

• Resource list from the University of Kansas Medical Center: Blindness / Visual Impairment
  http://www.kumc.edu/gec/support/visual.html

Clinical Information from GeneReviews

• X-Linked Congenital Stationary Night Blindness
  https://www.ncbi.nlm.nih.gov/books/NBK1245

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28x-linked+congenital+stationary+night+blindness%5BTIAB%5D%29+OR+%28x-linked+csnb%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22+AND+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

• NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A
  http://omim.org/entry/310500

• NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 2A
  http://omim.org/entry/300071

Medical Genetics Database from MedGen

• X-Linked Csnb
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14609846
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1771890/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9662400

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11062471

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10900517

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301423

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12397430

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11062472

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9662399

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