Autosomal dominant congenital stationary night blindness

Autosomal dominant 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 and distinguishing objects in low light (night blindness). For example, they are not able to identify road signs at night and some people cannot see stars in the night sky. Affected individuals have normal daytime vision and typically do not have other vision problems related to this disorder.

The night blindness associated with this condition is congenital, which means it is present from birth. This vision impairment tends to remain stable (stationary); it does not worsen over time.

Frequency

Autosomal dominant congenital stationary night blindness is likely a rare disease; however, its prevalence is unknown.

Causes

Mutations in the RHO, GNAT1, or PDE6B gene cause autosomal dominant congenital stationary night blindness. The proteins produced from these genes are necessary for normal vision, particularly in low-light conditions. These proteins are found in specialized light receptor cells in the retina called rods. Rods transmit visual signals from the eye to the brain when light is dim.

The RHO gene provides instructions for making a protein called rhodopsin, which is turned on (activated) by light entering the eye. Rhodopsin then attaches (binds) to and activates the protein produced from the GNAT1 gene, alpha (α)-transducin. The α-transducin protein then triggers the activation of a protein called cGMP-PDE, which is made up of multiple parts (subunits) including a subunit produced from the PDE6B gene. Activated cGMP-PDE triggers a series of chemical reactions that create electrical signals. These signals are transmitted from rod cells to the brain, where they are interpreted as vision.

Mutations in the RHO, GNAT1, or PDE6B gene disrupt the normal signaling that occurs within rod cells. As a result, the rods cannot effectively transmit signals to the brain, leading to a lack of visual perception in low light.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.
Other Names for This Condition

- adCSNB
- CSNBAD
- night blindness, congenital stationary, autosomal dominant

Diagnosis & Management

Genetic Testing Information

- What is genetic testing? [primer/testing/genetictesting]
- Genetic Testing Registry: Congenital stationary night blindness [https://www.ncbi.nlm.nih.gov/gtr/conditions/C0339535/]

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov [https://clinicaltrials.gov/ct2/results?cond=%22autosomal+dominant+congenital+stationary+night+blindness%22+OR+%22night+blindness%2C+congenital+stationary%22+OR+%22congenital+stationary+night+blindness%22]

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Vision--Night Blindness [https://medlineplus.gov/ency/article/003039.htm]
- Health Topic: Vision Impairment and Blindness [https://medlineplus.gov/visionimpairmentandblindness.html]

Educational Resources

- MalaCards: autosomal dominant congenital stationary night blindness [https://www.malacards.org/card/autosomal_dominant_congenital_stationary_night_blindness]
- Orphanet: Congenital stationary night blindness [https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=215]
- The University of Arizona: Congenital Stationary Night Blindness CSNBAD1 [https://disorders.eyes.arizona.edu/disorders/night-blindness-congenital-stationary-csnbad1]
• The University of Arizona: Congenital Stationary Night Blindness CSNBAD2
  https://disorders.eyes.arizona.edu/disorders/night-blindness-congenital-stationary-csnbad2

• The University of Arizona: Congenital Stationary Night Blindness CSNBAD3
  https://disorders.eyes.arizona.edu/disorders/night-blindness-congenital-stationary-csnbad3

Patient Support and Advocacy Resources

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

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

• Foundation Fighting Blindness (Canada)
  https://www.fightingblindness.ca/

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

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28congenital+stationary+night+blindness%5BTIAB%5D+AND+%28autosomal+dominant%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

• NIGHT BLINDNESS, CONGENITAL STATIONARY, AUTOSOMAL DOMINANT 1
  http://omim.org/entry/610445

• NIGHT BLINDNESS, CONGENITAL STATIONARY, AUTOSOMAL DOMINANT 2
  http://omim.org/entry/163500

• NIGHT BLINDNESS, CONGENITAL STATIONARY, AUTOSOMAL DOMINANT 3
  http://omim.org/entry/610444

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8075643

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20238025
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17584859

• Tsang SH, Woodruff ML, Jun L, Mahajan V, Yamashita CK, Pedersen R, Lin CS, Goff SP, Rosenberg T, Larsen M, Farber DB, Nusinowitz S. Transgenic mice carrying the H258N mutation in the gene encoding the beta-subunit of phosphodiesterase-6 (PDE6B) provide a model for human congenital stationary night blindness. Hum Mutat. 2007 Mar;28(3):243-54. 
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17044014
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2753261/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18487375

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