Autosomal recessive congenital stationary night blindness

Description

Autosomal recessive 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 may not be able to identify road signs at night or see stars in the night sky. They also often have other vision problems, including loss of sharpness (reduced acuity), nearsightedness (myopia), involuntary movements of the eyes (nystagmus), and eyes that do not look in the same direction (strabismus).

The vision problems associated with this condition are congenital, which means they are present from birth. They tend to remain stable (stationary) over time.

Frequency

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

Causes

Mutations in several genes can cause autosomal recessive congenital stationary night blindness. Each of these genes provide instructions for making proteins that are found in the retina. These proteins are involved in sending (transmitting) visual signals from cells called rods, which are specialized for vision in low light, to cells called bipolar cells, which relay the signals to other retinal cells. This signaling is an essential step in the transmission of visual information from the eyes to the brain.

Mutations in two genes, *GRM6* and *TRPM1*, cause most cases of this condition. These genes provide instructions for making proteins that are necessary for bipolar cells to receive and relay signals. Mutations in other genes involved in the same bipolar cell signaling pathway are likely responsible for a small percentage of cases of autosomal recessive congenital stationary night blindness.

Gene mutations that cause autosomal recessive congenital stationary night blindness disrupt the transmission of visual signals between rod cells and bipolar cells or interfere with the bipolar cells' ability to pass on these signals. As a result, visual information received by rod cells cannot be effectively transmitted to the brain, leading to difficulty seeing in low light. The cause of the other vision problems associated with this condition

is unclear. It has been suggested that the mechanisms that underlie night blindness can interfere with other visual systems, causing myopia, reduced visual acuity, and other impairments.

Some people with autosomal recessive congenital stationary night blindness have no identified mutation in any of the known genes. The cause of the disorder in these individuals is unknown.

Learn more about the genes associated with Autosomal recessive congenital stationary night blindness

- GRM6
- TRPM1

Additional Information from NCBI Gene:

- CABP4
- GPR179
- LRIT3
- SLC24A1

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Autosomal recessive complete congenital stationary night blindness
- Autosomal recessive incomplete congenital stationary night blindness

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Cone-rod synaptic disorder, congenital nonprogressive (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4041558/)
- Genetic Testing Registry: Congenital stationary night blindness 1B (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850362/)
- Genetic Testing Registry: Congenital stationary night blindness 1C (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750747/)

- Genetic Testing Registry: Congenital stationary night blindness 1D (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151193/)
- Genetic Testing Registry: Congenital stationary night blindness 1E (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3281215/)
- Genetic Testing Registry: Congenital stationary night blindness 1F (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3554399/)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1B; CSNB1B (https://omim.org/entry/257270)
- CONE-ROD SYNAPTIC DISORDER, CONGENITAL NONPROGRESSIVE; CRSD (https://omim.org/entry/610427)
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1E; CSNB1E (https://omim.org/entry/614565)
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1F; CSNB1F (https://omim.org/entry/615058)
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1C; CSNB1C (https://omim.org/entry/613216)
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1D; CSNB1D (https://omim.org/entry/613830)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28congenital+stationary+night+blindness%5BTIAB%5D%29+AND+%28autosomal+recessive%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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