adCSNB

Subjects: Genetics & Heredity

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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.

genetic conditions

1. Introduction

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.

2. Frequency

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

3. 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.

3.1. The genes associated with Autosomal dominant congenital stationary night blindness

- GNAT1
- PDE6B
- RHO

4. Inheritance

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.

5. Other Names for This Condition

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

References

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