Night Blindness, Congenital Stationary, Type 1C

Alternative Names
CSNB1C
CSNB, Complete, Autosomal Recessive

Record Category
Disease phenotype

WHO-ICD
Diseases of the eye and adnexa> Visual disturbances and blindness.

Incidence per 100,000 Live Births
Unknown

OMIM Number
613216

Mode of Inheritance
Autosomal recessive

Gene Map Locus
15q13.3

Description

Congenital stationary night blindness (CSNB) is a group of non-progressive retinal disorders characterized by an abnormal dark-adaptation curve, poor visual acuity, nystagmus, myopia (ranging from low to high), strabismus, and fundus abnormalities. There are two forms of CSNB, complete (CSNB1) and incomplete (CSNB2), according to the degree of rod function, measured by dark adaptometry or electroretinogram (ERG). The b-wave responses in patients with CSNB1C are severely deficient, but the a-waves are normal.

CSNB can be inherited as X linked recessive, autosomal recessive, or autosomal dominant patterns. Autosomal recessive congenital stationary night blindness is rare disease. However, the prevalence is unknown.

Molecular Genetics

CSNB1C is inherited as an autosomal recessive trait and caused by mutations in the TRPM1 (located on 15q13-q14). This gene codes for a calcium ion channel protein that is necessary for bipolar cells to receive and relay signals in the retina. At least 35 different mutations have been identified that cause disruption in the channel and prevent it from reaching the bipolar cell membrane and from relaying visual signals.

Epidemiology in the Arab World

Saudi Arabia
Al Oreany et al., (2016) described 20-year-old twin brothers with congenital stationary night blindness (CSNB). Both brothers presented with small amplitude pendular horizontal nystagmus, high myopia, hypoplastic titled discs and negative full field ERG with no measurable rod response. Next-generation sequencing identified a homozygous 1-bp deletion in exon 18 of the TRPM1 gene in both brothers. The parents were consanguineous and were both heterozygous carriers for the mutation.

References
Al Oreany AA, Al Hadlaq A1, Schatz P. Congenital stationary night blindness with hypoplastic discs, negative electroretinogram and thinning of the inner nuclear layer. Graefes Arch Clin Exp Ophthalmol. 2016;254(10):1951-1956. PMID: 27084085

Related CTGA Records
Transient Receptor Potential Cation Channel, Subfamily M, Member

External Links
https://ghr.nlm.nih.gov/condition/autosomal-recessive-congenital-stationary-night-blindness
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=215

Contributors
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