Transient Receptor Potential Cation Channel, Subfamily M, Member

Alternative Names
TRPM1
Melastatin 1
MLSN1

Record Category
Gene locus

WHO-ICD
N.B.: Classification not applicable to gene loci.

Incidence per 100,000 Live Births
N/A to gene loci

OMIM Number
603576

Mode of Inheritance
N/A

Gene Map Locus
15q13.3

Description
The TRPM1 gene codes for a member of the transient receptor potential (TRP) channel family that is important in cellular and somatosensory perception. The encoded protein is expressed in the bipolar cells and the melanocytes. The TRPM1 protein acts as a channel that transports cations in and out of bipolar cells. TRPM1 channels are involved in the pathway that is used to see in low light. When the TRPM1 channel is open, it allows cations to flow in and out of bipolar cells, which prevent visual signals from being sent in the bright-light conditions. While in the low-light conditions, the TRPM1 channels are triggered by the visual signals from rod cells to close, which causes visual signals to be transmitted. The channel may also play a role in Ca2+-dependent signaling related to melanocyte proliferation, differentiation, and/or survival.

Defects in this protein are the cause of congenital stationary night blindness type 1C (CSNB1), an autosomal recessive non-progressive retinal disorder, characterized by impaired night vision, nystagmus and myopia. Mutations in TRPM1 have been reported to account for at least half of the autosomal recessive CSNB1 cases in the Caucasian and Japanese population.

Molecular Genetics
The TRPM1 gene was mapped to the long arm of chromosome 15 (15q13.3), and consists of 27 coding exons spanning approximately 160 kb. The initiation codon in exon 3 results in a 1,533 residue protein that is predicted to contain six transmembrane domains. In addition, a SNP in the 5'UTR sequence can generate an additional initiation codon in exon 2, resulting in an isoform with 70 additional N-terminal amino acids. This allele is seen more frequently in lighter skinned individuals, raising the possibility of a link with melanoma.

At least 35 different mutations have been identified in patients with congenital stationary night blindness type 1C accounting for about half of the cases. These mutations disrupt 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. Using next-generation sequencing (NGS) in both brothers, a homozygous 1-bp deletion (c.2394delC) in exon 18 of the TRPM1 gene was identified. This mutation leads to a frameshift and subsequent formation of a premature stop codon (p.Thr799Profs*110), resulting in a truncated TRPM1 protein. The parents were consanguineous and both were found to be heterozygous carriers for the c.2394delC 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

Night Blindness, Congenital Stationary, Type 1C

External Links

https://ghr.nlm.nih.gov/gene/TRPM1
http://www.genecards.org/cgi-bin/carddisp.pl?gene=TRPM1

Contributors

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