PDE6B gene

phosphodiesterase 6B

Normal Function

The *PDE6B* gene provides instructions for making a protein that is one part (the beta subunit) of a protein complex called cGMP-PDE. This complex is found in specialized light receptor cells called rods. As part of the light-sensitive tissue at the back of the eye (the retina), rods transmit visual signals from the eye to the brain specifically in low-light conditions.

When light enters the eye, a series of rod cell proteins are turned on (activated), including cGMP-PDE. When cGMP-PDE is active, molecules called GMP within the rod cell are broken down, which triggers channels on the cell membrane to close. The closing of these channels results in the transmission of signals to the brain, which are interpreted as vision.

Health Conditions Related to Genetic Changes

Autosomal dominant congenital stationary night blindness

At least one mutation in the *PDE6B* gene has been found to cause autosomal dominant congenital stationary night blindness, which is characterized by the inability to see in low light. This mutation changes the protein building block (amino acid) histidine to the amino acid asparagine at position 258 in the beta subunit (written as His258Asp or H258N). This change impairs the normal function of the cGMP-PDE complex, causing it to be constantly turned on (constitutively active). Because the cGMP-PDE complex is always active, the signals that rod cells send to the brain are constantly occurring, even in bright light. Visual information from rod cells is then perceived by the brain as not meaningful, resulting in night blindness.

Retinitis pigmentosa

MedlinePlus Genetics provides information about Retinitis pigmentosa

Other Names for This Gene

- GMP-PDE beta
- PDE6B_HUMAN

- PDEB
- phosphodiesterase 6B, cGMP-specific, rod, beta
- rod cGMP-phosphodiesterase beta-subunit
- rod cGMP-specific 3',5'-cyclic phosphodiesterase subunit beta

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PDE6B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5158[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28PDE6B%5BTIAB%5D%29+OR+%28%28cGMP-PDE+beta%5BTIAB%5D%29+OR+%28PDEB%5BTIAB%5D%29+OR+%28rod+cGMP-phosphodiesterase+beta-subunit%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%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

- PHOSPHODIESTERASE 6B; PDE6B (https://omim.org/entry/180072)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5158)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PDE6B[gene])

References

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- Muradov KG, Granovsky AE, Artemyev NO. Mutation in rod PDE6 linked tocongenital stationary night blindness impairs the enzyme inhibition by itsgamma-subunit. Biochemistry. 2003 Mar 25;42(11):3305-10. doi: 10.1021/bi027095x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12641462)
- 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

carryingthe H258N mutation in the gene encoding the beta-subunit of phosphodiesterase-6(PDE6B) provide a model for human congenital stationary night blindness. HumMutat. 2007 Mar;28(3):243-54. doi: 10.1002/humu.20425. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17044014) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2753261/)

Genomic Location

The *PDE6B* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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