PDE6C Gene

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1. Introduction

The *PDE6C* gene provides instructions for making one part (called the alpha-prime subunit) of an enzyme called cone-specific phosphodiesterase. This enzyme is found exclusively in light-detecting (photoreceptor) cells called cones, which are located in a specialized tissue at the back of the eye known as the retina. Cones provide vision in bright light (daylight vision), including color vision. Other photoreceptor cells, called rods, provide vision in low light (night vision).

When light enters the eye, it stimulates specialized pigments in photoreceptor cells. This stimulation triggers a series of chemical reactions that produce an electrical signal, which is interpreted by the brain as vision. This process is called phototransduction. Cone-specific phosphodiesterase carries out one of the reactions in this process. Specifically, the enzyme converts a molecule called cGMP to another molecule, 5'-GMP, in cones. This conversion causes certain channels on the cell membrane to close. The closing of these channels triggers the transmission of visual signals to the brain.

2. Health Conditions Related to Genetic Changes

2.1. Achromatopsia

At least 19 mutations in the *PDE6C* gene have been found to cause the vision disorder achromatopsia. These mutations underlie a relatively small percentage of cases of complete achromatopsia, a form of the disorder characterized by a total lack of color vision and other vision problems that are present from early infancy. *PDE6C* gene mutations have also been identified in a few individuals with incomplete achromatopsia, a milder form of the disorder associated with limited color vision.

The *PDE6C* gene mutations associated with achromatopsia affect the function of the alpha-prime subunit. The mutations that underlie complete achromatopsia essentially eliminate the activity of cone-specific phosphodiesterase. Impairment of this enzyme disrupts the process of phototransduction in cones; rods are typically unaffected. Because cones are needed for color vision, people with complete achromatopsia can see only black, white, and shades of gray. They also have other vision problems related to malfunctioning cones, including reduced sharpness (low visual acuity), an increased sensitivity to light (photophobia), and involuntary back-and-forth eye movements (nystagmus).

Mutations in the *PDE6C* gene that reduce but do not eliminate the activity of cone-specific phosphodiesterase cause incomplete achromatopsia. People with incomplete achromatopsia have similar but less severe vision problems than people with complete achromatopsia, and they retain some color vision.

2.2. More About This Health Condition

Cone-rod dystrophy

3. Other Names for This Gene

- ACHM5
- cGMP phosphodiesterase 6C
- COD4
- cone cGMP-specific 3',5'-cyclic phosphodiesterase subunit alpha'

- PDEA2
- phosphodiesterase 6C, cGMP-specific, cone, alpha prime

References

1. Chang B, Grau T, Dangel S, Hurd R, Jurklies B, Sener EC, Andreasson S, DollfusH, Baumann B, Bolz S, Artemyev N, Kohl S, Heckenlively J, Wissinger B. Ahomologous genetic basis of the murine cpfl1 mutant and human achromatopsial inked to mutations in the PDE6C gene. Proc Natl Acad Sci U S A. 2009 Nov17;106(46):19581-6. doi: 10.1073/pnas.0907720106.
2. Grau T, Artemyev NO, Rosenberg T, Dollfus H, Haugen OH, Cumhur Sener E,Jurklies B, Andreasson S, Kernstock C, Larsen M, Zrenner E, Wissinger B, Kohl S. Decreased catalytic activity and altered activation properties of PDE6C mutantsassociated with autosomal recessive achromatopsia. Hum Mol Genet. 2011 Feb15;20(4):719-30. doi: 10.1093/hmg/ddq517.
3. Kohl S, Jägle H, Wissinger B, Zobor D. Achromatopsia. 2004 Jun 24 [updated2018 Sep 20]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University ofWashington, Seattle; 1993-2020. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1418/
4. Thiadens AA, den Hollander AI, Roosing S, Nabuurs SB, Zekveld-Vroon RC, CollinRW, De Baere E, Koenekoop RK, van Schooneveld MJ, Strom TM, van Lith-VerhoevenJJ, Lotery AJ, van Moll-Ramirez N, Leroy BP, van den Born LI, Hoyng CB, CremersFP, Klaver CC. Homozygosity mapping reveals PDE6C mutations in patients withearly-onset cone photoreceptor disorders. Am J Hum Genet. 2009 Aug;85(2):240-7.doi: 10.1016/j.ajhg.2009.06.016.