

PDE6C gene

phosphodiesterase 6C

Normal Function

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.

Health Conditions Related to Genetic Changes

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.

Cone-rod dystrophy

MedlinePlus Genetics provides information about Cone-rod dystrophy

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PDE6C ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5146\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5146[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PDE6C%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%5D>)

Catalog of Genes and Diseases from OMIM

- PHOSPHODIESTERASE 6C; PDE6C (<https://omim.org/entry/600827>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5146>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PDE6C\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PDE6C[gene]))

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Genomic Location

The *PDE6C* gene is found on chromosome 10 (<https://medlineplus.gov/genetics/chromosome/10/>).

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