

## 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

MedlinePlus Genetics provides information about Achromatopsia

#### 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>)

### 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]))

## References

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## Genomic Location

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

**Last updated January 1, 2015**