

## RHO gene

rhodopsin

### Normal Function

The *RHO* gene provides instructions for making a protein called rhodopsin. This protein is necessary for normal vision, particularly in low-light conditions. Rhodopsin 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 provide vision in low light. Other light receptor cells in the retina, called cones, are responsible for vision in bright light.

The rhodopsin protein is attached (bound) to a molecule called 11-cis retinal, which is a form of vitamin A. When light hits this molecule, it activates rhodopsin and sets off a series of chemical reactions that create electrical signals. These signals are transmitted to the brain, where they are interpreted as vision.

### Health Conditions Related to Genetic Changes

#### Autosomal dominant congenital stationary night blindness

At least four mutations in the *RHO* gene have been found to cause autosomal dominant congenital stationary night blindness, which is characterized by a loss of vision in low light that remains stable (stationary) over time. Unlike retinitis pigmentosa (described below), autosomal dominant congenital stationary night blindness does not affect daytime vision.

The *RHO* gene mutations responsible for autosomal dominant congenital stationary night blindness cause the rhodopsin protein to be constantly turned on (constitutively active). Because the protein no longer needs light to be activated, 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.

Researchers are uncertain why some constitutively activating mutations in the *RHO* gene cause congenital stationary night blindness and others result in the more severe vision loss associated with retinitis pigmentosa.

#### Retinitis pigmentosa

More than 150 mutations in the *RHO* gene have been identified in people with retinitis

pigmentosa. *RHO* gene mutations account for 20 to 30 percent of all cases of autosomal dominant retinitis pigmentosa, which is thought to be the most common form of the disorder. Rarely, mutations in the *RHO* gene cause autosomal recessive retinitis pigmentosa. However, this form of the disorder usually results from mutations in other genes.

Most of the *RHO* gene mutations responsible for retinitis pigmentosa alter the folding or transport of the rhodopsin protein. A few mutations cause rhodopsin to be constitutively activated instead of being activated in response to light. Studies suggest that altered versions of rhodopsin interfere with essential cell functions, causing rods to self-destruct (undergo apoptosis). Because rods are essential for vision under low-light conditions, the loss of these cells leads to progressive night blindness in people with retinitis pigmentosa.

Retinitis pigmentosa is also associated with a gradual loss of cone cells, which normally provide vision in bright light. The death of cone cells leads to tunnel vision and ultimately blindness in many affected individuals. It is unclear how mutations in the *RHO* gene affect the function and survival of cone cells.

### Other Names for This Gene

- CSNBAD1
- MGC138309
- MGC138311
- OPN2
- OPSD\_HUMAN
- opsin 2, rod pigment
- opsin-2
- RP4

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of RHO ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6010\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6010[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RHO%5BTI%5D%29+OR+%28rhodopsin%5BTI%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+360+days%22%5Bdp%5D%29%29%29>)

## Catalog of Genes and Diseases from OMIM

- RHODOPSIN; RHO (<https://omim.org/entry/180380>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6010>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RHO\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RHO[gene]))

## **References**

- Dryja TP, McGee TL, Reichel E, Hahn LB, Cowley GS, Yandell DW, Sandberg MA, Berson EL. A point mutation of the rhodopsin gene in one form of retinitis pigmentosa. *Nature*. 1990 Jan 25;343(6256):364-6. doi: 10.1038/343364a0. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/2137202>)
- McAlear SD, Kraft TW, Gross AK. 1 rhodopsin mutations in congenital nightblindness. *Adv Exp Med Biol*. 2010;664:263-72. doi: 10.1007/978-1-4419-1399-9\_30. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20238025>)
- Mendes HF, van der Spuy J, Chapple JP, Cheetham ME. Mechanisms of cell death in rhodopsin retinitis pigmentosa: implications for therapy. *Trends Mol Med*. 2005 Apr;11(4):177-85. doi: 10.1016/j.molmed.2005.02.007. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15823756>)
- Morris MB, Dastmalchi S, Church WB. Rhodopsin: structure, signal transduction and oligomerisation. *Int J Biochem Cell Biol*. 2009 Apr;41(4):721-4. doi: 10.1016/j.biocel.2008.04.025. Epub 2008 Aug 3. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18692154>)
- Palczewski K, Kumasaka T, Hori T, Behnke CA, Motoshima H, Fox BA, Le Trong I, Teller DC, Okada T, Stenkamp RE, Yamamoto M, Miyano M. Crystal structure of rhodopsin: A G protein-coupled receptor. *Science*. 2000 Aug 4;289(5480):739-45. doi: 10.1126/science.289.5480.739. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10926528>)
- Palczewski K. G protein-coupled receptor rhodopsin. *Annu Rev Biochem*. 2006;75:743-67. doi: 10.1146/annurev.biochem.75.103004.142743. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16756510>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1560097/>)
- Saliba RS, Munro PM, Luthert PJ, Cheetham ME. The cellular fate of mutant rhodopsin: quality control, degradation and aggresome formation. *J Cell Sci*. 2002 Jul 15;115(Pt 14):2907-18. doi: 10.1242/jcs.115.14.2907. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12082151>)
- Smith SO. Structure and activation of the visual pigment rhodopsin. *Annu Rev Biophys*. 2010;39:309-28. doi: 10.1146/annurev-biophys-101209-104901. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20192770>)
- Zeitz C, Gross AK, Leifert D, Kloeckener-Gruissem B, McAlear SD, Lemke J, Neidhardt J, Berger W. Identification and functional characterization of a

novelrhodopsin mutation associated with autosomal dominant CSNB. Invest Ophthalmol VisSci. 2008 Sep;49(9):4105-14. doi: 10.1167/iovs.08-1717. Epub 2008 May 16. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18487375>)

## **Genomic Location**

The *RHO* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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