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.

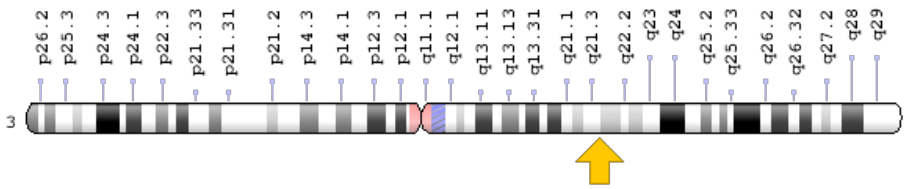
Most of the *RHO* gene mutations responsible for retinitis pigmentosa alter the folding or transport of the rhodopsin protein. A few mutations cause rhodopsin to be constitutively activated instead of being activated in response to light. Studies suggest that altered versions of rhodopsin interfere with 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.

Chromosomal Location

Cytogenetic Location: 3q22.1, which is the long (q) arm of chromosome 3 at position 22.1

Molecular Location: base pairs 129,528,639 to 129,535,344 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CSNBAD1
- MGC138309
- MGC138311
- OPN2
- OPSD_HUMAN
- opsin-2
- opsin 2, rod pigment
- RP4

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Rhodopsin, a Specialized 7TM Receptor, Absorbs Visible Light
  https://www.ncbi.nlm.nih.gov/books/NBK22541/#A4605
- Neuroscience (second edition, 2001): Phototransduction
  https://www.ncbi.nlm.nih.gov/books/NBK10806/
- Webvision: The Organization of the Retina and Visual System: Activation of Rod Phototransduction Cascade (figure)
  https://www.ncbi.nlm.nih.gov/books/NBK52768/figure/FuPhototran.F6/?report=objectonly

Clinical Information from GeneReviews

- Nonsyndromic Retinitis Pigmentosa Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1417

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

Catalog of Genes and Diseases from OMIM

- RHODOPSIN
  http://omim.org/entry/180380

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_RHO.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RHO%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10012
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6010
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6010

- RetNet: Summaries of Genes and Loci Causing Retinal Diseases
  https://sph.uth.edu/retnet/sum-dis.htm
- UniProt
  https://www.uniprot.org/uniprot/P08100

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