RPE65 gene

retinoid isomerohydrolase RPE65

Normal Function

The *RPE65* gene provides instructions for making a protein that is essential for normal vision. The RPE65 protein is produced in a thin layer of cells at the back of the eye called the retinal pigment epithelium (RPE). This cell layer supports and nourishes the retina, which is the light-sensitive tissue that lines the back of the eye. The RPE layer is essential for capturing light and vision.

The RPE65 protein is involved in a multi-step process called the visual cycle (also called the retinoid or vitamin A cycle), which converts light entering the eye into electrical signals that are transmitted to the brain. When light hits photosensitive pigments in the retina, it changes a molecule called 11-cis retinal (a form of vitamin A) to another molecule called all-trans retinal. This conversion triggers a series of chemical reactions that create electrical signals.

The RPE65 protein is a key enzyme in this cycle as it converts all-trans retinal to 11-cis retinol. Other enzymes then produce 11-cis retinal, so that the visual cycle can begin again and capture light.

Health Conditions Related to Genetic Changes

Leber congenital amaurosis

Many variants (also called mutations) in the *RPE65* gene have been found to cause Leber congenital amaurosis. This condition is an eye disorder that primarily affects the retina. People with this disorder typically have severe visual impairment beginning at birth or shortly afterward. Variants in the *RPE65* gene account for 6 to 16 percent of all cases of this condition.

RPE65 gene variants lead to a partial or total loss of RPE65 protein function. As a result, all-trans retinal cannot be converted back to 11-cis retinal, and excess all-trans retinal builds up in the retinal pigment epithelium. These abnormalities block the visual cycle, which leads to severe visual impairment beginning very early in life in Leber congenital amaurosis.

Fundus albipunctatus

MedlinePlus Genetics provides information about Fundus albipunctatus

Retinitis pigmentosa

MedlinePlus Genetics provides information about Retinitis pigmentosa

Other Names for This Gene

- all-trans-retinyl-palmitate hydrolase
- BCO3
- LCA2
- mRPE65
- p63
- RBP-binding membrane protein
- rd12
- retinal pigment epithelium specific protein 65
- retinal pigment epithelium-specific 65 kDa protein
- retinal pigment epithelium-specific protein 65kDa
- retinitis pigmentosa 20 (autosomal recessive)
- retinoid isomerohydrolase
- retinol isomerase
- RP20
- RPE65_HUMAN
- sRPE65

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RPE65 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6121[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28RPE65%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- RETINOID ISOMEROHYDROLASE RPE65; RPE65 (https://omim.org/entry/180069

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Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6121)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RPE65[gene])

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Genomic Location

The *RPE65* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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