RPE65

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1. 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 RPE65 protein is involved in a multi-step process called the visual 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 then helps convert all-trans retinal back to 11-cis retinal so the visual cycle can begin again.

2. Health Conditions Related to Genetic Changes

2.1. Leber congenital amaurosis

More than 30 mutations in the *RPE65* gene have been found to cause Leber congenital amaurosis. Mutations in this gene account for 6 to 16 percent of all cases of this condition.

RPE65 gene mutations 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.

2.2. Other disorders

More than 20 mutations in the *RPE65* gene have been identified in people with another eye disorder called retinitis pigmentosa. This condition is characterized by progressive vision loss caused by the gradual degeneration of light-sensing cells in the retina. The first sign of retinitis pigmentosa is usually a loss of night vision, which often becomes apparent in childhood. Over a period of years, the disease progresses to disrupt side (peripheral) vision and central vision. It may eventually lead to blindness.

The *RPE65* gene mutations that cause retinitis pigmentosa disrupt RPE65 protein function, which leads to vision loss by impairing the visual cycle. Mutations in this gene appear to be an uncommon cause of retinitis pigmentosa; these genetic changes are responsible for only a small percentage of all cases.

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

The entry is from https://medlineplus.gov/genetics/gene/rpe65

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