PRPH2 Gene

Subjects: Genetics & Heredity
Contributor: Karina Chen

peripherin 2

genes

1. Normal Function

The *PRPH2* gene (also known as *RDS*) provides instructions for making a protein called peripherin 2. This protein plays an important role in normal vision. Peripherin 2 is found in the retina, the light-sensitive tissue that lines the back of the eye. This protein is essential for the normal function of specialized cells called photoreceptors that detect light and color. Within these cells, peripherin 2 is involved in the formation and stability of structures that contain light-sensing pigments.

2. Health Conditions Related to Genetic Changes

2.1. Vitelliform macular dystrophy

Mutations in the *PRPH2* gene are responsible for some cases of adult-onset vitelliform macular dystrophy. Several mutations have been identified in people with this disorder, most of which change a single protein building block (amino acid) in peripherin 2. These mutations alter the protein's structure or lead to the production of an abnormally short, nonfunctional version of the protein. When peripherin 2 is altered or missing, photoreceptors break down (degenerate) over time. This loss of photoreceptors underlies the retinal abnormalities and progressive vision loss characteristic of vitelliform macular dystrophy. It is unclear why *PRPH2* mutations affect only central vision in people with this disorder.

2.2. Other retinal dystrophies

Mutations in the *PRPH2* gene cause a variety of other retinal disorders. Each of these conditions involves a slow degeneration of photoreceptor cells, leading to progressive vision loss. A total of more than 100 mutations in the *PRPH2* gene have been identified. Many of these mutations cause autosomal dominant retinitis pigmentosa, an eye disease that first disrupts night vision and side (peripheral) vision and eventually may result in blindness. *PRPH2* mutations also cause a group of retinal disorders called pattern dystrophies of the retinal pigment epithelium. These disorders typically begin in mid-adulthood and are characterized by an abnormal buildup of pigment in cells underlying the retina.

Some *PRPH2* mutations can cause different eye disorders in affected members of the same family. For example, researchers have reported a family with retinitis pigmentosa, pattern dystrophy of the retinal pigment epithelium, and retinitis punctata albescens (an eye disorder similar to retinitis pigmentosa) in different individuals with the same *PRPH2* mutation. It is unclear why mutations in this gene cause such a wide range of retinal abnormalities.

3. Other Names for This Gene

- CACD2
- peripherin 2 (retinal degeneration, slow)
- peripherin 2, homolog of mouse
- peripherin, photoreceptor type
- PRPH2_HUMAN
- RDS
- retinal degeneration slow protein
- retinal degeneration, slow
- Tetraspanin-22
- TSPAN22

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