CRB1 Gene

Subjects: Genetics & Heredity
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crumbs 1, cell polarity complex component

genes

1. Normal Function

The *CRB1* gene provides instructions for making a protein that plays an essential role in normal vision. This protein is found in the brain and the retina, which is the specialized tissue at the back of the eye that detects light and color.

In the retina, the CRB1 protein appears to be critical for the normal development of light-sensing cells called photoreceptors. Studies suggest that this protein is part of a group (complex) of proteins that help determine the structure and orientation of photoreceptors. The CRB1 protein may also be involved in forming connections between different types of cells in the retina.

2. Health Conditions Related to Genetic Changes

2.1. Leber Congenital Amaurosis

More than 50 mutations in the *CRB1* gene have been found to cause Leber congenital amaurosis. Mutations in this gene account for 9 to 13 percent of all cases of this condition.

Most of the *CRB1* gene mutations responsible for Leber congenital amaurosis lead to an abnormally short, nonfunctional version of the CRB1 protein or significantly reduce the amount of this protein produced in cells. A shortage of the CRB1 protein disrupts the early development of the retina. The retina becomes unusually thick and does not develop the normal layered structure. These changes cause severe visual impairment beginning very early in life.

2.2. Cone-Rod Dystrophy

Cone-rod dystrophy

2.3. Retinitis Pigmentosa

Retinitis pigmentosa

2.4. Other Disorders

At least 35 mutations in the *CRB1* 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 photoreceptors in the retina. *CRB1* gene mutations cause several uncommon forms of retinitis pigmentosa that are differentiated by their specific retinal changes.

The *CRB1* gene mutations that cause retinitis pigmentosa lead to a partial or total loss of CRB1 protein function. A shortage of normal CRB1 protein impairs the development of the retina and leads to the progressive degeneration of photoreceptors.

It is unclear why some people with *CRB1* gene mutations have severe, early visual impairment associated with Leber congenital amaurosis, and other people experience more gradual vision loss and other eye problems associated with retinitis pigmentosa. Researchers suspect that other genetic factors may modify the effects of *CRB1* gene mutations to influence the severity of these conditions.

3. Other Names for This Gene

- CRUM1_HUMAN
- crumbs family member 1, photoreceptor morphogenesis associated
- crumbs homolog 1
- crumbs homolog 1 (Drosophila)
- LCA8
- RP12

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