

CRB1 Gene

Subjects: Genetics & Heredity

Contributor: Vicky Zhou

crumbs 1, cell polarity complex component

Keywords: 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

References

1. den Hollander AI, Davis J, van der Velde-Visser SD, Zonneveld MN, Pierrottet CO, Koenekoop RK, Kellner U, van den Born LI, Heckenlively JR, Hoyng CB, Handford PA, Roepman R, Cremers FP. CRB1 mutation spectrum in inherited retinal dystrophies. Hum Mutat. 2004 Nov;24(5):355-69. Review.
2. den Hollander AI, Heckenlively JR, van den Born LI, de Kok YJ, van der Velde-Visser SD, Kellner U, Jurkies B, van Schooneveld MJ, Blankenagel A, Rohrschneider K, Wissinger B, Cruysberg JR, Deutman AF, Brunner HG, Apfelstedt-Sylla E, Hoyng CB, Cremers FP. Leber congenital amaurosis and retinitis pigmentosa with Coats-like exudative vasculopathy are associated with mutations in the crumbs homologue 1 (CRB1) gene. Am J Hum Genet. 2001 Jul;69(1):198-203. Nov;69(5):1160.
3. den Hollander AI, Roepman R, Koenekoop RK, Cremers FP. Leber congenital amaurosis: genes, proteins and disease mechanisms. Prog Retin Eye Res. 2008 Jul;27(4):391-419. doi: 10.1016/j.preteyeres.2008.05.003. Review.
4. den Hollander AI, ten Brink JB, de Kok YJ, van Soest S, van den Born LI, van Driel MA, van de Pol DJ, Payne AM, Bhattacharya SS, Kellner U, Hoyng CB, Westerveld A, Brunner HG, Bleeker-Wagemakers EM, Deutman AF, Heckenlively JR, Cremers FP, Bergen AA. Mutations in a human homologue of Drosophila crumbs cause retinitis pigmentosa (RP12). Nat Genet. 1999 Oct;23(2):217-21.
5. Gosens I, den Hollander AI, Cremers FP, Roepman R. Composition and function of the Crumbs protein complex in the mammalian retina. Exp Eye Res. 2008 May;86(5):713-26. doi: 10.1016/j.exer.2008.02.005.
6. Jacobson SG, Cideciyan AV, Aleman TS, Pianta MJ, Sumaroka A, Schwartz SB, Smilko EE, Milam AH, Sheffield VC, Stone EM. Crumbs homolog 1 (CRB1) mutations result in a thick human retina with abnormal lamination. Hum Mol Genet. 2003 May 1;12(9):1073-8.
7. Lotery AJ, Jacobson SG, Fishman GA, Weleber RG, Fulton AB, Namperumalsamy P, Héon E, Levin AV, Grover S, Rosenow JR, Kopp KK, Sheffield VC, Stone EM. Mutations in the CRB1 gene cause Leber congenital amaurosis. Arch Ophthalmol. 2001 Mar;119(3):415-20.
8. Richard M, Roepman R, Aartsen WM, van Rossum AG, den Hollander AI, Knust E, Wijnholds J, Cremers FP. Towards understanding CRUMBS function in retinal dystrophies. Hum Mol Genet. 2006 Oct 15;15 Spec No 2:R235-43. Review.
9. van de Pavert SA, Kantardzhieva A, Malysheva A, Meuleman J, Versteeg I, Levelt C, Klooster J, Geiger S, Seeliger MW, Rashbass P, Le Bivic A, Wijnholds J. Crumbs homologue 1 is required for maintenance of photoreceptor cell polarization and adhesion during light exposure. J Cell Sci. 2004 Aug 15;117(Pt 18):4169-77.

Retrieved from <https://encyclopedia.pub/entry/history/show/12307>