

## CRB1 gene

crumbs cell polarity complex component 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.

### Health Conditions Related to Genetic Changes

#### Leber congenital amaurosis

Many variants (also called mutations) in the *CRB1* gene have been found to cause Leber congenital amaurosis. Leber congenital amaurosis 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 *CRB1* gene account for 9 to 13 percent of all cases of this condition.

Most of the *CRB1* gene variants 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.

#### Cone-rod dystrophy

MedlinePlus Genetics provides information about Cone-rod dystrophy

#### Retinitis pigmentosa

MedlinePlus Genetics provides information about Retinitis pigmentosa

## Other Names for This Gene

- CRUM1\_HUMAN
- crumbs family member 1, photoreceptor morphogenesis associated
- crumbs homolog 1
- crumbs homolog 1 (Drosophila)
- LCA8
- RP12

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of CRB1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23418\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23418[geneid]))

### Scientific Articles on PubMed

- PubMed ([https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CRB1%5BTIAB%5D%29+OR+%28crumbs+homolog\\*+1%5BTIAB%5D%29%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%29%29%29](https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CRB1%5BTIAB%5D%29+OR+%28crumbs+homolog*+1%5BTIAB%5D%29%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%29%29%29))

### Catalog of Genes and Diseases from OMIM

- RETINITIS PIGMENTOSA 12; RP12 (<https://omim.org/entry/600105>)
- CRUMBS CELL POLARITY COMPLEX COMPONENT 1; CRB1 (<https://omim.org/entry/604210>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/23418>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CRB1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CRB1[gene]))

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## Genomic Location

The *CRB1* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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