

## RPGR gene

retinitis pigmentosa GTPase regulator

### Normal Function

The *RPGR* gene provides instructions for making a protein that is essential for normal vision. Although the protein's function is not well understood, studies suggest that it plays an important role in cell structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of many types of cells. They are involved in cell movement and many different chemical signaling pathways. Cilia are also necessary for the perception of sensory input, including hearing, smell, and vision.

Several different versions (isoforms) of the RPGR protein are produced from the *RPGR* gene. One version contains a segment known as the ORF15 exon. This version of the RPGR protein is active (expressed) predominantly in the retina, which is the light-sensitive tissue at the back of the eye. Specifically, the ORF15-containing isoform is found in the retina's specialized light receptor cells (photoreceptors). Researchers suspect that this isoform may help maintain photoreceptors by regulating the function of cilia. Other isoforms of the RPGR protein are expressed in other parts of the body, where they are probably also involved in cilia function.

### Health Conditions Related to Genetic Changes

#### Retinitis pigmentosa

More than 300 mutations in the *RPGR* gene have been found to cause the X-linked form of retinitis pigmentosa. This condition primarily affects males, causing night blindness in early childhood followed by progressive daytime vision loss. *RPGR* gene mutations account for about 70 percent of all cases of X-linked retinitis pigmentosa.

Most of the mutations responsible for X-linked retinitis pigmentosa occur in the ORF15 exon of the RPGR protein. These mutations usually result in an abnormally short, malfunctioning protein. Changes in the structure of the RPGR protein likely disrupt the normal function of cilia in photoreceptor cells. However, it is unclear how these changes lead to the gradual loss of photoreceptors and resulting vision problems that are characteristic of retinitis pigmentosa.

#### Cone-rod dystrophy

Mutations in the *RPGR* gene can cause X-linked cone-rod dystrophy. The problems associated with this condition include a loss of visual sharpness (acuity), an increased sensitivity to light (photophobia), and impaired color vision. These vision problems worsen over time.

The genetic changes that cause X-linked cone-rod dystrophy likely disrupt the function of cilia in photoreceptor cells. It is unclear how these changes lead to the pattern of photoreceptor loss that results in cone-rod dystrophy.

### Primary ciliary dyskinesia

MedlinePlus Genetics provides information about Primary ciliary dyskinesia

### Other disorders

Although most *RPGR* gene mutations cause X-linked retinitis pigmentosa (described above), a few mutations in the ORF15 exon have been found in people with other retinal disorders. These include cone-rod dystrophy (described above), cone dystrophy, and atrophic macular degeneration. These retinal disorders are characterized by progressive vision abnormalities, although their signs and symptoms are distinct from retinitis pigmentosa.

Several additional *RPGR* gene mutations have been reported in people with a combination of retinitis pigmentosa and signs and symptoms affecting other parts of the body. In addition to progressive vision loss, affected individuals can have chronic respiratory and sinus infections, recurrent ear infections (otitis media), and hearing loss.

It is unclear why mutations in the *RPGR* gene can cause a variety of disorders. Studies suggest that certain mutations may disrupt the function of cilia in multiple tissues, including the inner ear and respiratory tract. Malfunctioning cilia in these tissues may underlie the hearing loss and respiratory abnormalities seen in some affected individuals. However, researchers are still working to determine how *RPGR* gene mutations cause specific abnormalities involving the retina and other parts of the body.

### **Other Names for This Gene**

- COD1
- CORDX1
- CRD
- PCDX
- retinitis pigmentosa 15
- retinitis pigmentosa 3 GTPase regulator
- RP15
- RP3
- RPGR\_HUMAN
- X-linked retinitis pigmentosa GTPase regulator

- XLRP3

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of RPGR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6103\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6103[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RPGR%5BTIAB%5D%29+OR+%28retinitis+pigmentosa+GTPase+regulator%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>)

### Catalog of Genes and Diseases from OMIM

- RETINITIS PIGMENTOSA, X-LINKED, AND SINORESPIRATORY INFECTIONS WITH OR WITHOUT DEAFNESS; RPSRDF (<https://omim.org/entry/300455>)
- RETINITIS PIGMENTOSA GTPase REGULATOR; RPGR (<https://omim.org/entry/312610>)
- CONE-ROD DYSTROPHY, X-LINKED, 1; CORDX1 (<https://omim.org/entry/304020>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6103>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RPGR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RPGR[gene]))

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## Genomic Location

The *RPGR* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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