RP2 gene

RP2 activator of ARL3 GTPase

Normal Function

The *RP2* gene provides instructions for making a protein that is essential for normal vision. The RP2 protein is active in cells throughout the body, including cells that make up the light-sensitive tissue at the back of the eye (the retina). However, the function of the RP2 protein is not well understood. Studies suggest that it may be involved in transporting proteins within the retina's specialized light receptor cells (photoreceptors). Its role in other types of cells is unknown.

Health Conditions Related to Genetic Changes

Retinitis pigmentosa

More than 70 mutations in the *RP2* gene have been identified in people with the X-linked form of retinitis pigmentosa. This condition primarily affects males, causing night blindness in early childhood followed by progressive daytime vision loss. *RP2* gene mutations account for 10 to 15 percent of all cases of X-linked retinitis pigmentosa.

Most mutations in the *RP2* gene lead to the production of an abnormally short version of the RP2 protein. A few mutations change single building blocks (amino acids) in the RP2 protein. These changes alter the structure and function of the protein, which probably disrupts the stability or maintenance of photoreceptor cells. A gradual loss of photoreceptors underlies the progressive vision loss characteristic of retinitis pigmentosa.

Other Names for This Gene

- DELXp11.3
- KIAA0215
- NM23-H10
- NME10
- protein XRP2
- retinitis pigmentosa 2 (X-linked recessive)
- TBCCD2

- XRP2
- XRP2_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RP2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6102[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RP2%5BTIAB%5D%29+OR+%28retinitis+pigmentosa+2%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- RP2 ACTIVATOR OF ARL3 GTPase; RP2 (https://omim.org/entry/300757)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6102)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RP2[gene])

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Genomic Location

The *RP2* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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