

## RS1 gene

retinoschisin 1

### Normal Function

The *RS1* gene provides instructions for making a protein called retinoschisin, which is found in the retina. The retina is a specialized light-sensitive tissue that lines the back of the eye. Retinoschisin attaches (binds) to the surface of specialized cells within the retina that detect light and color (photoreceptor cells). The protein also binds to bipolar cells, which relay light signals from photoreceptor cells to other retinal cells. Studies suggest that retinoschisin plays a role in the development and maintenance of the retina and its specialized cells. Retinoschisin is likely involved in the organization of cells in the retina by attaching cells together (cell adhesion).

### Health Conditions Related to Genetic Changes

#### X-linked juvenile retinoschisis

More than 220 mutations in the *RS1* gene have been found to cause X-linked juvenile retinoschisis. This disorder causes tiny splits (schisis) or tears to form in the retina, which results in progressive vision loss in males. Most of the *RS1* gene mutations change one protein building block (amino acid) in the retinoschisin protein, although many different types of mutations have been identified. Research suggests that the various mutations in the *RS1* gene can alter the 3-dimensional structure of the protein, impair the protein's ability to attach cells together (cell adhesion), cause misplacement of the protein within retinal cells, or prevent protein production. Changes in the retinoschisin protein function or production disrupt its role in the maintenance and organization of the retina. As a result, splitting or tearing of the retina can occur, causing the vision problems associated with X-linked juvenile retinoschisis.

### Other Names for This Gene

- retinoschisin
- retinoschisis (X-linked, juvenile) 1
- RS
- X-linked juvenile retinoschisis protein
- XLRS1
- XLRS1\_HUMAN

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of RS1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6247\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6247[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RS1%5BTIAB%5D%29+OR+%28retinoschisin%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>)

### Catalog of Genes and Diseases from OMIM

- RETINOSCHISIS 1, X-LINKED, JUVENILE; RS1 (<https://omim.org/entry/312700>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6247>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RS1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RS1[gene]))

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## Genomic Location

The *RS1* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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