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])

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])

References

- Kim DY, Mukai S. X-linked juvenile retinoschisis (XLRS): a review ofgenotype-phenotype relationships. Semin Ophthalmol. 2013 Sep-Nov;28(5-6):392-6.doi: 10.3109/08820538.2013.825299. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24138048)
- Molday RS, Kellner U, Weber BH. X-linked juvenile retinoschisis: clinicaldiagnosis, genetic analysis, and molecular mechanisms. Prog Retin Eye Res. 2012May;31(3):195-212. doi: 10.1016/j.preteyeres.2011.12.002. Epub 2012 Jan 3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22245536) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3334421/)
- Molday RS. Focus on molecules: retinoschisin (RS1). Exp Eye Res. 2007Feb;84(2):227-8. doi: 10.1016/j.exer.2005.12.013. Epub 2006 Apr 4. No abstractavailable. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16600216)
- Pimenides D, George ND, Yates JR, Bradshaw K, Roberts SA, Moore AT, Trump D. X-linked retinoschisis: clinical phenotype and RS1 genotype in 86 UK patients. JMed Genet. 2005 Jun;42(6):e35. doi: 10.1136/jmg.2004.029769. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15937075) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1736077/)
- Wu WW, Wong JP, Kast J, Molday RS. RS1, a discoidin domain-containing retinalcell adhesion protein associated with X-linked retinoschisis, exists as a

noveldisulfide-linked octamer. J Biol Chem. 2005 Mar 18;280(11):10721-30. doi:10.1074/jbc.M413117200. Epub 2005 Jan 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15644328)

- Yi J, Li S, Jia X, Xiao X, Wang P, Guo X, Zhang Q. Novel RS1 mutationsassociated with X-linked juvenile retinoschisis. Int J Mol Med. 2012Apr;29(4):644-8. doi: 10.3892/ijmm.2012.882. Epub 2012 Jan 10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22245991) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3573736/)

Genomic Location

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

Last updated March 1, 2015