

SKIV2L gene

Ski2 like RNA helicase

Normal Function

The *SKIV2L* gene provides instructions for making a protein whose function has not been confirmed. Based on its similarity to a protein in other organisms, researchers speculate that the SKIV2L protein acts as part of a group of proteins called the SKI complex. This complex is thought to be necessary for the function of another large protein complex known as the cytosolic exosome. Within cells, the cytosolic exosome helps to recognize and break down excess or abnormal messenger RNA (mRNA) molecules. mRNA is a chemical cousin of DNA that serves as the genetic blueprint for protein production. Studies suggest that the cytosolic exosome's role in getting rid of excess and abnormal mRNA is important for cell growth.

Health Conditions Related to Genetic Changes

Trichohepatoenteric syndrome

At least nine mutations in the *SKIV2L* gene have been found to cause trichohepatoenteric syndrome, a rare condition that affects many parts of the body. Its major signs and symptoms include chronic diarrhea starting in infancy, hair abnormalities, distinctive facial features, and liver disease. Mutations in this gene likely eliminate the function of the SKIV2L protein. Researchers hypothesize that a loss of this protein's function impairs the activity of the SKI complex and the cytosolic exosome. However, it is unknown how these changes could lead to chronic diarrhea and the other features of trichohepatoenteric syndrome.

Other Names for This Gene

- 170A
- DDX13
- helicase SKI2W
- helicase-like protein
- HLP
- SKI2
- SKI2 homolog, superkiller viralicidic activity 2-like

- SKI2W
- SKIV2
- SKIV2L1
- superkiller viralicidic activity 2-like (*S. cerevisiae*)
- THES2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SKIC2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6499\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6499[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SKI2W%5BTIAB%5D%29+OR+%28SKI2W%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

Catalog of Genes and Diseases from OMIM

- SKI2 SUBUNIT OF SUPERKILLER COMPLEX; SKIC2 (<https://omim.org/entry/600478>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6499>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SKI2L\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SKI2L[gene]))

References

- Fabre A, Charroux B, Martinez-Vinson C, Roquelaure B, Odul E, Sayar E, Smith H, Colomb V, Andre N, Hugot JP, Goulet O, Lacoste C, Sarles J, Royet J, Levy N, Badens C. SKIV2L mutations cause syndromic diarrhea, or trichohepatoenteric syndrome. *Am J Hum Genet.* 2012 Apr 6;90(4):689-92. doi:10.1016/j.ajhg.2012.02.009. Epub 2012 Mar 22. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22444670>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3322239/>)
- Fabre A, Martinez-Vinson C, Goulet O, Badens C. Syndromic diarrhea/Trichohepato-enteric syndrome. *Orphanet J Rare Dis.* 2013 Jan 9;8:5. doi: 10.1186/1750-1172-8-5. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23302111>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3560276/>)

- van Dijk EL, Schilders G, Pruijn GJ. Human cell growth requires a functional cytoplasmic exosome, which is involved in various mRNA decay pathways. RNA. 2007 Jul;13(7):1027-35. doi: 10.1261/rna.575107. Epub 2007 Jun 1. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17545563>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1894934/>)

Genomic Location

The *SKIV2L* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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