



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

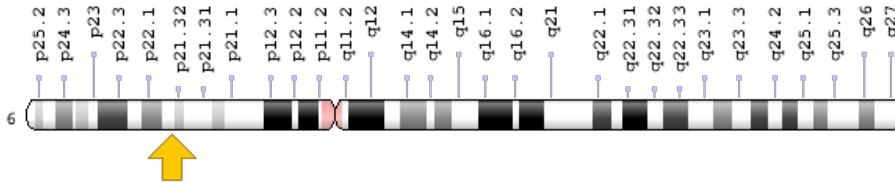
Trichohepatoenteric syndrome

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.

Chromosomal Location

Cytogenetic Location: 6p21.33, which is the short (p) arm of chromosome 6 at position 21.33

Molecular Location: base pairs 31,959,157 to 31,969,852 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 170A
- DDX13
- helicase-like protein
- helicase SKI2W
- HLP
- SKI2
- SKI2 homolog, superkiller viralicidic activity 2-like
- SKI2W
- SKIV2
- SKIV2L1
- superkiller viralicidic activity 2-like (*S. cerevisiae*)
- THES2

Additional Information & Resources

Educational Resources

- Genomes (second edition, 2002): Degradation of mRNAs
<https://www.ncbi.nlm.nih.gov/books/NBK21132/#A7399>
- Marie Curie Bioscience Database: Structural Components and Architectures of RNA Exosomes
<https://www.ncbi.nlm.nih.gov/books/NBK45033/>

Clinical Information from GeneReviews

- Trichohepatoenteric Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK475802>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SKIV2L%5BTIAB%5D%29+OR+%28SKI2W%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

Catalog of Genes and Diseases from OMIM

- SKI2-LIKE RNA HELICASE
<http://omim.org/entry/600478>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SKIV2L.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SKIV2L%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10898
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6499>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6499>
- UniProt
<https://www.uniprot.org/uniprot/Q15477>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/SKIV2L>

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