

UTP4 gene

UTP4 small subunit processome component

Normal Function

The *UTP4* gene provides instructions for making a protein called cirhin, whose precise function is unknown. This protein is found in many different tissues and organs. Within cells, cirhin is located in a structure called the nucleolus, which is a small region inside the nucleus where ribosomal RNA (rRNA) is produced. A chemical cousin of DNA, rRNA is a molecule that helps assemble protein building blocks (amino acids) into functioning proteins. Researchers believe that cirhin may play a role in processing rRNA.

Studies also suggest that cirhin interacts with other proteins, and it may function as part of a protein complex (a group of proteins that work together). The significance of these protein interactions is unknown.

Health Conditions Related to Genetic Changes

North American Indian childhood cirrhosis

At least one mutation in the *UTP4* gene has been found to cause North American Indian childhood cirrhosis, a chronic liver disease identified in a First Nation population in Quebec. The known mutation replaces the amino acid arginine with the amino acid tryptophan at protein position 565 (written as Arg565Trp or R565W). This genetic change likely alters the structure and function of cirhin. However, it is unclear why the effects of the *UTP4* gene mutation are limited to the liver or how they cause the progressive liver damage characteristic of this disorder.

Other Names for This Gene

- CIR1A_HUMAN
- CIRH1A
- cirhin
- cirrhosis, autosomal recessive 1A (cirhin)
- FLJ14728
- FLJ17146

- KIAA1988
- NAIC
- testis expressed gene 292
- TEX292

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of UTP4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=84916\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=84916[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CIRH1A%5BTIAB%5D%29+OR+%28UTP4%5BTIAB%5D%29%29+OR+%28cirhin%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

Catalog of Genes and Diseases from OMIM

- UTP4 SMALL SUBUNIT PROCESSOME COMPONENT; UTP4 (<https://omim.org/entry/607456>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/84916>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=UTP4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=UTP4[gene]))

References

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Genomic Location

The *UTP4* gene is found on chromosome 16 (<https://medlineplus.gov/genetics/chromosome/16/>).

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