

## TTC37 gene

tetratricopeptide repeat domain 37

### Normal Function

The *TTC37* 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 TTC37 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 25 mutations in the *TTC37* 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 TTC37 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

- KIAA0372
- Ski3
- SKI3 homolog
- tetratricopeptide repeat protein 37
- thespin
- TPR repeat protein 37
- tricho-hepatic-enteric syndrome protein

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of SKIC3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9652\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9652[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28TTC37%5BTIAB%5D%29+OR+%28Ski3%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

### Catalog of Genes and Diseases from OMIM

- SKI3 SUBUNIT OF SUPERKILLER COMPLEX; SKIC3 (<https://omim.org/entry/614589>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9652>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=TTC37\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=TTC37[gene]))

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## Genomic Location

The *TTC37* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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