TRPS1 gene
transcriptional repressor GATA binding 1

Normal Function

The TRPS1 gene provides instructions for making a protein that regulates the activity of many other genes. The TRPS1 protein is found within the cell nucleus where it interacts with specific regions of DNA to turn off (repress) gene activity. Based on this role, the TRPS1 protein is called a transcription factor. Research suggests that the TRPS1 protein plays a role in regulating genes that control the growth of bone and cartilage, a tough but flexible tissue that makes up much of the skeleton during early development.

Health Conditions Related to Genetic Changes

Trichorhinophalangeal syndrome type I

More than 130 mutations in the TRPS1 gene have been found to cause trichorhinophalangeal syndrome type I (TRPS I). TRPS I is a condition that causes bone and joint malformations; distinctive facial features; and abnormalities of the skin, hair, teeth, sweat glands, and nails. The mutations that cause this condition affect one copy of the TRPS1 gene in each cell.

Some of these mutations change single protein building blocks (amino acids) and have a "dominant-negative" effect, which means that the altered protein produced from one copy of the TRPS1 gene interferes with the function of the normal protein produced from the other copy of the gene. Other mutations add or delete small amounts of genetic material or alter the way the gene's instructions are used to make the protein. These changes have a "loss-of-function" effect and result in the production of a protein that cannot enter the cell nucleus where it is needed to bind to DNA and repress gene activity.

As a result of the protein's decreased ability to repress gene activity, particularly genes that regulate bone and cartilage growth, people with TRPS1 gene mutations develop abnormal bones in the fingers and toes, joint abnormalities, distinctive facial features, and other signs and symptoms of TRPS I.

Trichorhinophalangeal syndrome type II

The TRPS1 gene is located in a region of chromosome 8 that is deleted in people with trichorhinophalangeal syndrome type II (TRPS II). TRPS II is a condition that causes bone and joint malformations; distinctive facial features; intellectual disability; and abnormalities of the skin, hair, teeth, sweat glands, and nails. As a result of this deletion, affected individuals are missing one copy of the TRPS1 gene in each cell and TRPS1 protein production is reduced by half. Researchers believe that
this reduction in TRPS1 protein impairs the regulation of certain genes that control bone and cartilage growth, which contributes to short stature, abnormal bones in the fingers and toes, joint abnormalities, and distinctive facial features in people with TRPS II. The deletion of other genes near the TRPS1 gene likely contributes to the additional features of this condition.

Chromosomal Location

Cytogenetic Location: 8q23.3, which is the long (q) arm of chromosome 8 at position 23.3

Molecular Location: base pairs 115,408,496 to 115,669,637 on chromosome 8 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• GC79
• LGCR
• TRPS1_HUMAN
• zinc finger transcription factor TRPS1

Additional Information & Resources

Clinical Information from GeneReviews

• Trichorhinophalangeal Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK425926

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28TRPS1%5BTIAB%5D%29+OR+%28LGCR%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22+AND+5Bdp%5D
Catalog of Genes and Diseases from OMIM

- **ZINC FINGER TRANSCRIPTION FACTOR TRPS1**  
  http://omim.org/entry/604386

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
  http://atlasgeneticsoncology.org/Genes/GC_TRPS1.html
- ClinVar  
  https://www.ncbi.nlm.nih.gov/clinvar?term=TRPS1%5Bgene%5D
- HGNC Gene Symbol Report  
- Monarch Initiative  
  https://monarchinitiative.org/gene/NCBIGene:7227
- NCBI Gene  
- UniProt  
  https://www.uniprot.org/uniprot/Q9UHF7

Sources for This Summary

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