TRIP11 gene
thyroid hormone receptor interactor 11

Normal Function

The TRIP11 gene provides instructions for making a protein known as Golgi microtubule-associated protein 210 (GMAP-210). This protein is found in the Golgi apparatus, a cell structure in which newly produced proteins are modified so they can carry out their functions. Studies suggest that the GMAP-210 protein helps to maintain the structure of the Golgi apparatus, and it may also be involved in the transport of certain proteins out of cells.

Although the GMAP-210 protein is found throughout the body, researchers suspect that it may have a particularly important role in cells called chondrocytes in the developing skeleton. Chondrocytes give rise to cartilage, a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears.

Health Conditions Related to Genetic Changes

Achondrogenesis

At least nine mutations in the TRIP11 gene have been found to cause a form of achondrogenesis known as type 1A or the Houston-Harris type. This rare disorder of bone development is characterized by extremely short limbs, a narrow chest, short ribs that fracture easily, and a lack of normal bone formation (ossification) in the skull, spine, and pelvis. Serious health problems result from these abnormalities, and infants with achondrogenesis usually die before or soon after birth.

The TRIP11 gene mutations associated with achondrogenesis type 1A lead to the production of a nonfunctional version of the GMAP-210 protein or prevent the cell from producing any of this protein. Studies suggest that a shortage of GMAP-210 activity alters the structure and function of the Golgi apparatus, which impairs protein modification. Chondrocytes appear to be particularly sensitive to these changes, and malfunction of the Golgi apparatus in these cells likely underlies the problems with bone formation in achondrogenesis type 1A.
**Chromosomal Location**

Cytogenetic Location: 14q32.12, which is the long (q) arm of chromosome 14 at position 32.12

Molecular Location: base pairs 91,965,991 to 92,040,134 on chromosome 14 (Homo sapiens Updated Annotation Release 109.20191205, GRCh38.p13) (NCBI)

[Diagram of chromosome 14 with location highlighted]

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- ACG1A
- CEV14
- clonal evolution-related gene on chromosome 14 protein
- GMAP-210
- Golgi-associated microtubule-binding protein 210
- Golgi-microtubule-associated protein of 210 kDa
- thyroid receptor-interacting protein 11
- TR-interacting protein 11
- TRIP-11
- TRIP230
- TRIPB_HUMAN

**Additional Information & Resources**

**Educational Resources**

- Madame Curie Bioscience Database: The Golgi Apparatus
  https://www.ncbi.nlm.nih.gov/books/NBK6268/
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TRIP11%5BTIAB%5D%29+OR+%28thyroid+hormone+receptor+interactor+11%5BTIAB%5D%29+OR+%28%28GMAP-210%5BTIAB%5D%29+OR+%28TRIP-11%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- THYROID HORMONE RECEPTOR INTERACTOR 11
  http://omim.org/entry/604505

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/CEV14ID96.html
- ClinVar
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9321
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q15643

Sources for This Summary

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