TRAPPC2 gene
trafficking protein particle complex 2

Normal Function

The TRAPPC2 gene provides instructions for producing the protein sedlin, which is found in cells throughout the body. Sedlin is part of a large group of proteins called the trafficking protein particle (TRAPP) complex, which plays a role in the transport of proteins between cell compartments (organelles). Sedlin is thought to be located between two organelles, the endoplasmic reticulum and the Golgi apparatus. The endoplasmic reticulum is involved in protein processing and transport, and the Golgi apparatus modifies newly produced proteins.

Research shows that sedlin is required for transporting large proteins from the endoplasmic reticulum to the Golgi apparatus. For example, sedlin is needed to move large molecules called procollagens out of the endoplasmic reticulum so they can be processed further by the Golgi apparatus. Later, procollagens are altered by enzymes outside the cell to create smaller mature collagen proteins, which strengthen and support connective tissues, such as skin, bone, cartilage, tendons, and ligaments.

Health Conditions Related to Genetic Changes

X-linked spondyloepiphyseal dysplasia tarda

More than 50 mutations in the TRAPPC2 gene have been found to cause X-linked spondyloepiphyseal dysplasia tarda. This condition impairs bone growth and occurs almost exclusively in males, usually appearing between ages 6 and 10. Almost all mutations result in a nonfunctional sedlin protein. As a result, large proteins, including procollagen, cannot be transported out of the endoplasmic reticulum. A lack of procollagen transport reduces the amount of mature collagen in cells, which impairs the development of bones, cartilage, and other connective tissues. It is likely that this disruption in bone development leads to many of the signs and symptoms of X-linked spondyloepiphyseal dysplasia tarda, although it is unclear why the skeletal problems do not appear until later in childhood.
Chromosomal Location

Cytogenetic Location: Xp22.2, which is the short (p) arm of the X chromosome at position 22.2

Molecular Location: base pairs 13,712,242 to 13,734,635 on the X chromosome (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MBP-1 interacting protein-2A
- MIP-2A
- SEDL
- sedlin
- SEDT
- TPPC2_HUMAN
- TRS20
- ZNF547L

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience: Regulation and Coordination of Intracellular Trafficking: An Overview
  https://www.ncbi.nlm.nih.gov/books/NBK7285/

Clinical Information from GeneReviews

- X-Linked Spondyloepiphyseal Dysplasia Tarda
  https://www.ncbi.nlm.nih.gov/books/NBK1145
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28TRAPPC2%5BALL%5D%29+OR+%28%28%28SED1%5BTIAB%5D%29+OR+%28sedlin%5BTIAB%5D%29+AND+%28%28SEDT%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- TRACKING PROTEIN PARTICLE COMPLEX, SUBUNIT 2
  http://omim.org/entry/300202

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TRAPPC2%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6399

- NCBI Gene

- UniProt: TPC2A_HUMAN
  https://www.uniprot.org/uniprot/P0DI81

- UniProt: TPC2B_HUMAN
  https://www.uniprot.org/uniprot/P0DI82

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11595175

- OMIM: TRACKING PROTEIN PARTICLE COMPLEX, SUBUNIT 2
  http://omim.org/entry/300202

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23019651
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3471527/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23876379

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/24841781

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