TRAPPC2 gene

trafficking protein particle complex subunit 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.

Other Names for This Gene

- MBP-1 interacting protein-2A
- MIP-2A

- SEDL
- sedlin
- SEDT
- TPPC2_HUMAN
- TRS20
- ZNF547L

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TRAPPC2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6399[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TRAPPC2%5BALL%5D%29+OR+%28%28SEDL%5BTIAB%5D%29+OR+%28sedlin%5BTIAB%5D%29+OR+%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; TRAPPC2 (https://omim.org/entry/300202)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6399)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TRAPPC2[gene])

References

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Genomic Location

The *TRAPPC2* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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