CRTAP gene
cartilage associated protein

Normal Function

The CRTAP gene provides instructions for making a protein called cartilage associated protein. While the specific function of this protein is not known, it plays an important role in normal bone development. Cartilage associated protein works with two other proteins, leprecan and cyclophilin B, as part of a complex that helps process certain forms of collagen. Collagens are proteins that provide strength, support, and the ability to stretch (elasticity) to many body tissues.

The complex containing cartilage associated protein modifies a protein building block (amino acid) called proline in collagen molecules. This modification, which is known as proline 3-hydroxylation, appears to be critical for the normal folding and assembly of collagen. It also may be important for releasing collagen molecules into the spaces around cells (the extracellular matrix). The secretion of collagen from cells is necessary for the proper formation of connective tissues, such as bones, tendons, and cartilage, that form the body’s supportive framework.

Health Conditions Related to Genetic Changes

Osteogenesis imperfecta

At least five mutations in the CRTAP gene are responsible for a rare type of osteogenesis imperfecta that is usually classified as type VII. Several of these mutations prevent cells from producing any cartilage associated protein. Without this protein, bones and other connective tissues do not form properly, leading to a very severe form of the disorder. Another mutation in the CRTAP gene greatly reduces the amount of cartilage associated protein produced, which disrupts the normal formation of collagen. This genetic change causes less severe signs and symptoms of osteogenesis imperfecta.
Chromosomal Location

Cytogenetic Location: 3p22.3, which is the short (p) arm of chromosome 3 at position 22.3

Molecular Location: base pairs 33,113,958 to 33,147,773 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cartilage-associated protein
- CASP
- CRTAP_HUMAN

Additional Information & Resources

Educational Resources

- Howard Hughes Medical Institute: Genetic Mutation Explains Form of Brittle Bone Disease (October 20, 2006)

  https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3551

  https://www.ncbi.nlm.nih.gov/books/NBK21582/

  https://www.ncbi.nlm.nih.gov/books/NBK9874/?rendertype=figure&id=A2050

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CRTAP%5BTIAB%5D%29+OR+%28cartilage+associated+protein%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22+AND+journal%5Bdp%5D
Catalog of Genes and Diseases from OMIM

• CARTILAGE-ASSOCIATED PROTEIN
  http://omim.org/entry/605497

Research Resources

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

• HGNC Gene Symbol Report

• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:10491

• NCBI Gene

• UniProt
  https://www.uniprot.org/uniprot/O75718

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17192541

• OMIM: CARTILAGE-ASSOCIATED PROTEIN
  http://omim.org/entry/605497

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12110407

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17630507

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17055431

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10702664

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12110406

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