COL9A2 gene

collagen type IX alpha 2 chain

Normal Function

The *COL9A2* gene provides instructions for making part of a large molecule called type IX collagen. Collagens are a family of proteins that strengthen and support connective tissues, such as skin, bone, cartilage, tendons, and ligaments. In particular, type IX collagen is an important component of cartilage, which is 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.

Type IX collagen is made up of three proteins that are produced from three distinct genes: one α1(IX) chain, which is produced from the *COL9A1* gene, one α2(IX) chain, which is produced from the *COL9A2* gene, and one α3(IX) chain, which is produced from the *COL9A3* gene. Type IX collagen is more flexible than other types of collagen molecules and is closely associated with type II collagen. Researchers believe that the flexible nature of type IX collagen allows it to act as a bridge that connects type II collagen with other cartilage components. Studies have shown that type IX collagen also interacts with the proteins produced from the *MATN3* and *COMP* genes.

Health Conditions Related to Genetic Changes

Multiple epiphyseal dysplasia

At least five mutations in the *COL9A2* gene have been shown to cause dominant multiple epiphyseal dysplasia, a disorder of cartilage and bone development that primarily affects the ends of the long bones in the arms and legs (epiphyses). All of these mutations disrupt how genetic information is spliced together to make the blueprint for producing the α2(IX) chain. These mutations, called splice-site mutations, change one DNA building block (nucleotide) near an area of the gene called exon 3. These mutations in the *COL9A2* gene result in the deletion of 12 protein building blocks (amino acids) from the α2(IX) chain. It is not known how mutations in *COL9A2* cause the signs and symptoms of dominant multiple epiphyseal dysplasia.

Stickler syndrome

MedlinePlus Genetics provides information about Stickler syndrome

Intervertebral disc disease

MedlinePlus Genetics provides information about Intervertebral disc disease

Other Names for This Gene

- alpha 2 type IX collagen
- CO9A2_HUMAN
- collagen IX, alpha-2 polypeptide
- collagen type IX alpha 2
- collagen, type IX, alpha 2
- EDM2
- epiphyseal dysplasia, multiple 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COL9A2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1298[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28COL9A2%5BTIAB%5D%29+OR+%28EDM2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE IX, ALPHA-2; COL9A2 (https://omim.org/entry/120260)
- INTERVERTEBRAL DISC DISEASE; IDD (https://omim.org/entry/603932)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1298)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=COL9A2[gene])

References

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Genomic Location

The *COL9A2* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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