

COL9A3 gene

collagen type IX alpha 3 chain

Normal Function

The *COL9A3* 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.

Type IX collagen is made up of three proteins that are produced from three distinct genes: one $\alpha 1(\text{IX})$ chain, which is produced from the *COL9A1* gene, one $\alpha 2(\text{IX})$ chain, which is produced from the *COL9A2* gene, and one $\alpha 3(\text{IX})$ 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 three mutations in the *COL9A3* gene have been shown to cause dominant multiple epiphyseal dysplasia. All of these mutations disrupt how genetic information is spliced together to make the blueprint for producing the $\alpha 3(\text{IX})$ 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 *COL9A3* gene result in the deletion of 12 protein building blocks (amino acids) from the $\alpha 3(\text{IX})$ chain. It is not known how mutations in *COL9A3* cause the signs and symptoms of dominant multiple epiphyseal dysplasia.

All identified mutations in type IX collagen delete a portion of the COL3 domain, which suggests that this region has an important function. Mutations may affect the ability of type IX collagen to fold correctly or interact with other cartilage components.

Intervertebral disc disease

MedlinePlus Genetics provides information about Intervertebral disc disease

Stickler syndrome

MedlinePlus Genetics provides information about Stickler syndrome

Other Names for This Gene

- alpha 3 type IX collagen
- CO9A3_HUMAN
- collagen type IX alpha 3
- collagen, type IX, alpha 3
- DJ885L7.4.1
- EDM3
- FLJ90759
- IDD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COL9A3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1299\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1299[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28COL9A3%5BTIAB%5D%29+OR+%28EDM3%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%29>)

Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE IX, ALPHA-3; COL9A3 (<https://omim.org/entry/120270>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1299>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COL9A3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COL9A3[gene]))

References

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(<https://pubmed.ncbi.nlm.nih.gov/11968079>)

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Genomic Location

The COL9A3 gene is found on chromosome 20 (<https://medlineplus.gov/genetics/chromosome/20/>).

Last updated February 1, 2008