



## COL9A1 gene

collagen type IX alpha 1 chain

### Normal Function

The *COL9A1* 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  $\alpha 1$ (IX) chain, which is produced from the *COL9A1* gene, one  $\alpha 2$ (IX) chain, which is produced from the *COL9A2* gene, and one  $\alpha 3$ (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

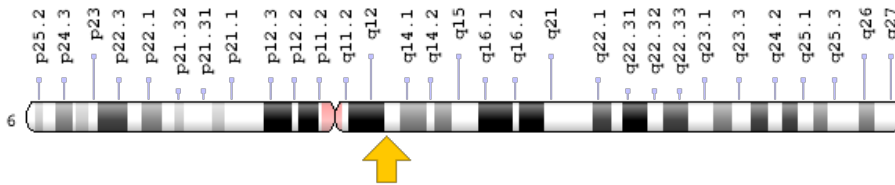
At least one mutation in the *COL9A1* gene has been found to cause dominant multiple epiphyseal dysplasia, a disorder of cartilage and bone development that primarily affects the ends of the long bones in the arms and legs (epiphyses). The identified mutation, called a splice-site mutation, involves the addition of one DNA building block (nucleotide) near an area of the gene called exon 8 (written as 1-bp ins, IVS8, T, +3). This mutation disrupts the way the gene's instructions are used to make the  $\alpha 1$ (IX) chain, resulting in a deletion of several protein building blocks (amino acids). It is not known how this mutation in *COL9A1* causes the signs and symptoms of dominant multiple epiphyseal dysplasia.

#### Stickler syndrome

## Chromosomal Location

Cytogenetic Location: 6q13, which is the long (q) arm of chromosome 6 at position 13

Molecular Location: base pairs 70,215,061 to 70,303,084 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- alpha 1 type IX collagen
- cartilage-specific short collagen
- collagen IX, alpha-1 polypeptide
- collagen type IX alpha 1
- collagen, type IX, alpha 1
- DJ149L1.1.2
- FLJ40263
- MED

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Collagens Are the Major Proteins of the Extracellular Matrix  
<https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3551>
- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix  
<https://www.ncbi.nlm.nih.gov/books/NBK21582/>

### Clinical Information from GeneReviews

- Multiple Epiphyseal Dysplasia, Dominant  
<https://www.ncbi.nlm.nih.gov/books/NBK1123>
- Stickler Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1302>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL9A1%5BTIAB%5D%29+OR+%28%28multiple+epiphyseal+dysplasia%5BTIAB%5D%29+OR+%28collagen+IX%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+360+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE IX, ALPHA-1  
<http://omim.org/entry/120210>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=COL9A1%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:2217](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2217)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:1297>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1297>
- UniProt  
<https://www.uniprot.org/uniprot/P20849>

### **Sources for This Summary**

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Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/COL9A1>

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