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 \( \alpha_1(IX) \) 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 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 \( \alpha_3(IX) \) 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 \( \alpha_3(IX) \) 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**

**Stickler syndrome**
Chromosomal Location

Cytogenetic Location: 20q13.33, which is the long (q) arm of chromosome 20 at position 13.33

Molecular Location: base pairs 62,817,050 to 62,841,159 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

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

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

  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=%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+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

• COLLAGEN, TYPE IX, ALPHA-3
  http://omim.org/entry/120270

Research Resources

• ClinVar

• HGNC Gene Symbol Report

• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1299

• NCBI Gene

• UniProt
  https://www.uniprot.org/uniprot/Q14050

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11968079

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301302

• OMIM: COLLAGEN, TYPE IX, ALPHA-3
  http://omim.org/entry/120270
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/24273071

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16586133 
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2267527/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16371896

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