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 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 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 α1(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

MedlinePlus Genetics provides information about Stickler syndrome

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

Tests Listed in the Genetic Testing Registry

- Tests of COL9A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1297[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; COL9A1 (https://omim.org/entry/120210)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1297)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=COL9A1[gene])

References

- Blumbach K, Niehoff A, Paulsson M, Zaucke F. Ablation of collagen IX and COMPdisrupts epiphyseal cartilage architecture. Matrix Biol. 2008 May;27(4):306-18. doi: 10.1016/j.matbio.2007.11.007. Epub 2007 Dec 3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18191556)
- Briggs MD, Chapman KL. Pseudoachondroplasia and multiple epiphyseal dysplasia:

mutation review, molecular interactions, and genotype to phenotype correlations. Hum Mutat. 2002 May;19(5):465-78. doi: 10.1002/humu.10066. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11968079)

- Briggs MD, Wright MJ, Mortier GR. Multiple Epiphyseal Dysplasia, AutosomalDominant. 2003 Jan 8 [updated 2019 Apr 25]. In: Adam MP, Feldman J, Mirzaa GM,Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R) [Internet]. Seattle (WA): University of Washington,Seattle; 1993-2024. Available from http://www.ncbi.nlm.nih.gov/books/NBK1123/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301302)
- Czarny-Ratajczak M, Lohiniva J, Rogala P, Kozlowski K, Perala M, Carter L,Spector TD, Kolodziej L, Seppanen U, Glazar R, Krolewski J, Latos-Bielenska A,Ala-Kokko L. A mutation in COL9A1 causes multiple epiphyseal dysplasia: furtherevidence for locus heterogeneity. Am J Hum Genet. 2001 Nov;69(5):969-80. doi:10.1086/324023. Epub 2001 Sep 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11565064) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1274373/)
- Nikopoulos K, Schrauwen I, Simon M, Collin RW, Veckeneer M, Keymolen K, VanCamp G, Cremers FP, van den Born LI. Autosomal recessive Stickler syndrome in twofamilies is caused by mutations in the COL9A1 gene. Invest Ophthalmol Vis Sci.2011 Jul 1;52(7):4774-9. doi: 10.1167/iovs.10-7128. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21421862)
- Van Camp G, Snoeckx RL, Hilgert N, van den Ende J, Fukuoka H, Wagatsuma M, Suzuki H, Smets RM, Vanhoenacker F, Declau F, Van de Heyning P, Usami S. A newautosomal recessive form of Stickler syndrome is caused by a mutation in theCOL9A1 gene. Am J Hum Genet. 2006 Sep;79(3):449-57. doi: 10.1086/506478. Epub2006 Jun 26. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16909383) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1559536/)

Genomic Location

The *COL9A1* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

Last updated January 1, 2013