

COL5A1 gene

collagen type V alpha 1 chain

Normal Function

The *COL5A1* gene provides instructions for making a component of type V collagen. Collagens are a family of proteins that strengthen and support many tissues in the body, including skin, ligaments, bones, tendons, and muscles.

A component of type V collagen called the pro- α 1(V) chain is produced from the *COL5A1* gene. Collagens begin as rope-like procollagen molecules that are each made up of three chains. Two combinations of chains can produce type V collagen: three pro- α 1(V) chains or two pro- α 1(V) chains and one pro- α 2(V) chain (which is produced from the *COL5A2* gene).

The triple-stranded procollagen molecules are processed by enzymes outside the cell to create mature collagen. The collagen molecules then arrange themselves into long, thin fibrils with another form of collagen, type I. Type V collagen regulates the width (diameter) of those fibrils. Studies suggest that type V collagen also controls the assembly of other types of collagen into fibrils in several tissues.

Health Conditions Related to Genetic Changes

Ehlers-Danlos syndrome

Mutations in the *COL5A1* gene cause a form of Ehlers-Danlos syndrome called the classical type. Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues that support the skin, bones, blood vessels, and many other organs and tissues. This form of the disorder is characterized by skin that is soft, highly stretchy (elastic), and fragile; abnormal scarring; and an unusually large range of joint movement (hypermobility). More than 100 *COL5A1* gene mutations have been identified in people with classical Ehlers-Danlos syndrome. The mutations affect one copy of the gene in each cell, reducing the amount of pro- α 1(V) chains that cells produce. As a result, fibrils containing type V and type I collagens in the skin and other tissues are disorganized and larger than usual. Researchers believe that the abnormal collagen weakens connective tissues throughout the body, which causes the signs and symptoms of classical Ehlers-Danlos syndrome.

Carpal tunnel syndrome

MedlinePlus Genetics provides information about Carpal tunnel syndrome

Keratoconus

MedlinePlus Genetics provides information about Keratoconus

Other Names for This Gene

- alpha 1 type V collagen preproprotein
- CO5A1_HUMAN
- collagen type V alpha 1
- collagen, type V, alpha 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COL5A1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1289\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1289[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28COL5A1%5BTIAB%5D%29+OR+%28%28collagen%5BTI%5D%29+AND+%28type+V%5BTI%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE V, ALPHA-1; COL5A1 (<https://omim.org/entry/120215>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1289>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COL5A1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COL5A1[gene]))

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Genomic Location

The *COL5A1* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

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