

## COL5A2 gene

collagen type V alpha 2 chain

### Normal Function

The *COL5A2* 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- $\alpha$ 2(V) chain is produced from the *COL5A2* 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- $\alpha$ 1(V) chains (produced from the *COL5A1* gene) or two pro- $\alpha$ 1(V) chains and one pro- $\alpha$ 2(V) chain.

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 *COL5A2* gene have been identified in a small number of people with 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). About 20 *COL5A2* gene mutations have been identified in people with classical Ehlers-Danlos syndrome. These mutations, which affect one copy of the gene in each cell, change the structure of the pro- $\alpha$ 2(V) chain. As a result, fibrils containing type V and type I collagens 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.

## Other Names for This Gene

- AB collagen
- CO5A2\_HUMAN
- collagen type V alpha 2
- collagen V, alpha-2 polypeptide
- collagen, fetal membrane, A polypeptide
- collagen, type V, alpha 2

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of COL5A2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1290\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1290[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28COL5A2%5BTIAB%5D%29+OR+%28AB+collagen%5BTIAB%5D%29+OR+%28%28collagen%5BTI%5D%29+AND+%28type+V%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE V, ALPHA-2; COL5A2 (<https://omim.org/entry/120190>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1290>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COL5A2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COL5A2[gene]))

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## Genomic Location

The COL5A2 gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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