

COL3A1 gene

collagen type III alpha 1 chain

Normal Function

The *COL3A1* gene provides instructions for making type III collagen. Collagens are a family of proteins that strengthen and support many tissues in the body. Type III collagen is found in the skin, lungs, intestinal walls, and the walls of blood vessels.

The components of type III collagen, called pro- α 1(III) chains, are produced from the *COL3A1* gene. Each molecule of type III procollagen is made up of three copies of this chain.

The triple-stranded, rope-like procollagen molecules are processed by enzymes outside the cell to create mature type III collagen. The collagen molecules then arrange themselves into long, thin fibrils that form stable interactions (cross-links) with one another and with other types of collagen in the spaces between cells. The cross-links result in the formation of very strong collagen fibers.

Health Conditions Related to Genetic Changes

Ehlers-Danlos syndrome

More than 500 mutations in the *COL3A1* gene have been found to cause a form of Ehlers-Danlos syndrome called the vascular 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. The vascular type can cause potentially life-threatening complications, including tearing (rupture) of blood vessels, intestines, and other organs. The mutations that cause this form of the disorder alter the structure and production of type III procollagen molecules. As a result, a large percentage of type III collagen molecules are assembled incorrectly, or the amount of type III collagen is greatly reduced. Researchers believe that these changes affect tissues that are normally rich in this type of collagen, such as the skin, blood vessel walls, and internal organs. An insufficient amount of type III collagen weakens connective tissues in these parts of the body, causing the signs and symptoms of the vascular type of Ehlers-Danlos syndrome.

Other Names for This Gene

- alpha 1 type III collagen
- CO3A1_HUMAN
- collagen III, alpha-1 polypeptide
- collagen type III alpha 1
- collagen, fetal
- collagen, type III, alpha 1
- collagen, type III, alpha 1 (Ehlers-Danlos syndrome type IV, autosomal dominant)
- EDS4A

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COL3A1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1281\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1281[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28COL3A1%5BTIAB%5D%29+OR+%28%28collagen%5BTI%5D%29+AND+%28type+III%5BTI%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Dp%5D>)

Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE III, ALPHA-1; COL3A1 (<https://omim.org/entry/120180>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1281>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COL3A1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COL3A1[gene]))

References

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Genomic Location

The *COL3A1* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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