

COL4A5 gene

collagen type IV alpha 5 chain

Normal Function

The *COL4A5* gene provides instructions for making one piece of a protein called collagen IV. Specifically, this gene makes the alpha5(IV) chain of collagen IV. This chain combines with two other types of alpha (IV) chains (the alpha3 and alpha4 chains) to make alpha345(IV) collagen molecules. These molecules attach to each other to form complex protein networks that make up a large portion of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues. Alpha345(IV) collagen networks play an especially important role in the basement membranes of the kidney, inner ear, and eye.

Health Conditions Related to Genetic Changes

Alport syndrome

Many variants (also called mutations) in the *COL4A5* gene can cause Alport syndrome, a condition characterized by kidney disease, hearing loss, and eye abnormalities. Most of these variants change single protein building blocks (amino acids) in a region where the alpha5(IV) collagen chain combines with other collagen IV chains. Other variants in the *COL4A5* gene severely decrease or prevent the production of alpha5(IV) chains.

These changes in the structure or amount of alpha5(IV) chains severely impair the formation of alpha345(IV) collagen networks in the basement membranes of the kidney, inner ear, and eye. In the kidney, other types of collagen build up in the basement membranes, eventually scarring the kidneys and leading to kidney failure. Variants in this gene can also lead to hearing loss and changes in the lens of the eye and the light-sensitive tissue at the back of the eye (retina).

Other Names for This Gene

- ASLN
- ATS
- CA54
- CO4A5_HUMAN
- collagen IV, alpha-5 polypeptide

- collagen of basement membrane, alpha-5 chain
- collagen type IV alpha 5
- collagen, type IV, alpha 5
- collagen, type IV, alpha 5 (Alport syndrome)

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COL4A5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1287\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1287[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28COL4A5%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+1800+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE IV, ALPHA-5; COL4A5 (<https://omim.org/entry/303630>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1287>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COL4A5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COL4A5[gene]))

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Genomic Location

The *COL4A5* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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