COL4A5 gene
collagen type IV alpha 5 chain

Normal Function

The \textit{COL4A5} gene provides instructions for making one component of type IV collagen, which is a flexible protein. Specifically, this gene makes the alpha5(IV) chain of type IV collagen. This chain combines with two other types of alpha (IV) chains (the alpha3 and alpha4 chains) to make a complete type IV collagen molecule. Type IV collagen molecules attach to each other to form complex protein networks. These networks make up a large portion of basement membranes, which are thin sheet-like structures that separate and support cells in many tissues. Type IV collagen alpha3-4-5 networks play an especially important role in the basement membranes of the kidney, inner ear, and eye.

Health Conditions Related to Genetic Changes

\textbf{Alport syndrome}

More than 400 mutations in the \textit{COL4A5} gene have been found to cause Alport syndrome. Most of these mutations change single protein building blocks (amino acids) in a region where the alpha5(IV) collagen chain combines with other type IV collagen chains. Other mutations in the \textit{COL4A5} gene severely decrease or prevent the production of alpha5(IV) chains. As a result, there is a serious deficiency of the type IV collagen alpha3-4-5 network in the basement membranes of the kidney, inner ear, and eye. In the kidney, other types of collagen accumulate in the basement membranes, eventually leading to scarring of the kidneys and kidney failure. Mutations in this gene can also lead to abnormal function in the inner ear, resulting in hearing loss.
Chromosomal Location

Cytogenetic Location: Xq22.3, which is the long (q) arm of the X chromosome at position 22.3

Molecular Location: base pairs 108,439,844 to 108,697,545 on the X chromosome (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

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

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK26810/?rendertype=figure&id=A3581
  https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3583
Clinical Information from GeneReviews

- Alport Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1207

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/303630

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_COL4A5.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=COL4A5%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1287
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P29400

Sources for This Summary

- OMIM: COLLAGEN, TYPE IV, ALPHA-5
  http://omim.org/entry/303630
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12105244
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14514738

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10752524

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301386

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10499074

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15021198

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12768082

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17396119

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