COL18A1 gene

collagen type XVIII alpha 1 chain

Normal Function

The *COL18A1* gene provides instructions for making a protein that forms collagen XVIII. Three COL18A1 proteins, called alpha 1 subunits, attach to each other to form collagen XVIII. Collagen XVIII is found in the basement membranes of tissues throughout the body. Basement membranes are thin, sheet-like structures that separate and support cells in these tissues.

There are three versions (isoforms) of the alpha 1 subunit of collagen XVIII, which form three different lengths of the collagen XVIII protein. The short version of collagen XVIII is found in basement membranes throughout the body, including several parts of the eye. The two longer versions are found primarily in the liver. In addition, a piece of collagen XVIII can be cut off to form the protein endostatin. Endostatin is able to block the formation of blood vessels (angiogenesis) and is known as an anti-angiogenic factor.

Little is known about the function of collagen XVIII, but it appears that all of the isoforms are involved in normal development of the eye.

Health Conditions Related to Genetic Changes

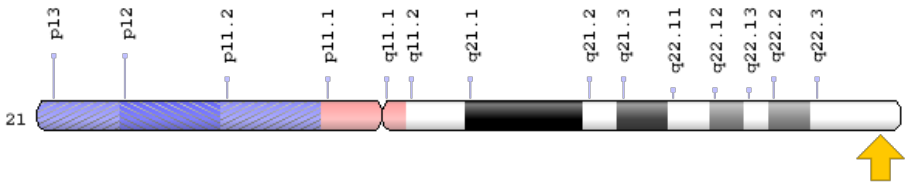
Knobloch syndrome

At least a dozen mutations in the *COL18A1* gene have been identified in people with Knobloch syndrome, a condition characterized by severe vision problems and a skull defect called an occipital encephalocele. Most *COL18A1* gene mutations lead to an abnormally short version of the genetic blueprint used to make the alpha 1 subunit of collagen XVIII. There are a few other mutations that replace single protein building blocks (amino acids) in the protein sequence. Although the process is unclear, the *COL18A1* gene mutations result in the loss of collagen XVIII. Most *COL18A1* gene mutations affect all isoforms of collagen XVIII, but at least one mutation affects only the short isoform. The loss of one or all isoforms of the collagen XVIII protein likely causes the signs and symptoms of Knobloch syndrome. It is unclear whether endostatin is involved in this condition.

Chromosomal Location

Cytogenetic Location: 21q22.3, which is the long (q) arm of chromosome 21 at position 22.3

Molecular Location: base pairs 45,405,165 to 45,513,720 on chromosome 21 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- antiangiogenic agent
- collagen alpha-1(XVIII) chain
- collagen type XVIII alpha 1
- collagen, type XVIII, alpha 1
- endostatin
- FLJ27325
- FLJ34914
- KNO
- KNO1
- KS
- MGC74745
- multi-functional protein MFP

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL18A1%5BTIAB%5D%29+OR+%28%28collagen+alpha-1++chain%5BTIAB%5D%29+OR+%28endostatin%5BTIAB%5D%29+OR+%28KNO%5BTIAB%5D%29+OR+%28KNO1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- COLLAGEN, TYPE XVIII, ALPHA-1
  http://omim.org/entry/120328

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_COL18A1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=COL18A1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2195
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:80781
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/80781
- UniProt
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