



TGFBI gene

transforming growth factor beta induced

Normal Function

The *TGFBI* gene provides instructions for making a protein called transforming growth factor beta induced (TGFBI). This protein is released (secreted) from cells and becomes part of the extracellular matrix, which is an intricate network that forms in the spaces between cells and provides structural support to tissues. The TGFBI protein is thought to play a role in the attachment of cells to one another (cell adhesion) and cell movement (migration). This protein is found in many tissues in the body, including the the clear, outer covering of the eye (the cornea).

Health Conditions Related to Genetic Changes

Lattice corneal dystrophy type I

At least 26 mutations in the *TGFBI* gene can cause lattice corneal dystrophy type I. This inherited eye condition is characterized by a lattice-like accumulation of proteins (called amyloid deposits) that form in the cornea. These deposits cloud the cornea and lead to vision impairment in affected individuals. The cornea is made up of several layers of tissue. In lattice corneal dystrophy type I, the deposits occur in the stromal layer of the cornea and contain altered TGFBI proteins.

The *TGFBI* gene mutations involved in lattice corneal dystrophy type I change single protein building blocks (amino acids) in the TGFBI protein. The most common mutation replaces the amino acid arginine with the amino acid cysteine at position 124 (written as Arg124Cys or R124C). When the condition is caused by this mutation, it is often called classic lattice corneal dystrophy type I. Altered TGFBI proteins abnormally clump together and form amyloid deposits. However, it is unclear how the changes caused by the gene mutations induce the protein to form these deposits.

Keratoconus

Other disorders

TGFBI gene mutations are involved in a number of other types of corneal dystrophy, including Groenouw corneal dystrophy, Avellino corneal dystrophy, lattice corneal dystrophy type IIIA, Reis-Bucklers corneal dystrophy, Thiel-Behnke corneal dystrophy, and epithelial basement membrane corneal dystrophy. Corneal dystrophies are all characterized by the accumulation of protein deposits in the cornea, which leads to vision impairment. These deposits form in various shapes and

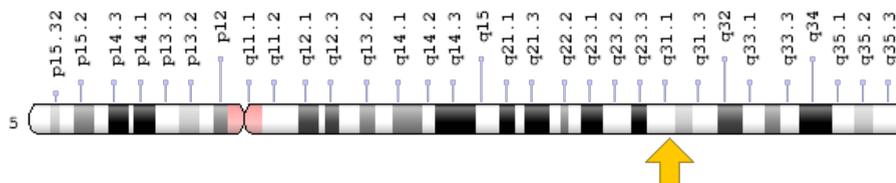
patterns and occur in different layers of the cornea, which help define the different types of corneal dystrophies.

The *TGFBI* gene mutations that cause these conditions change single amino acids in the TGFBI protein. Most mutations alter the amino acids found at positions 124 and 555 of the protein, although other amino acid changes can also occur. Certain mutations are strongly associated with particular corneal dystrophies. For instance, the mutation that replaces the amino acid arginine at position 124 with the amino acid histidine (written as Arg124His) most often causes Avellino corneal dystrophy. The mutation that replaces the amino acid arginine at position 124 with the amino acid lysine (written as Arg124Lys) most often causes Reis-Bucklers corneal dystrophy. The mutation that replaces the amino acid arginine at position 555 with the amino acid glutamine (written as Arg555Gln) most often causes Thiel-Behnke corneal dystrophy. And, the mutation that replaces the amino acid arginine at position 555 with the amino acid tryptophan (written as Arg555Trp) most often causes Groenouw corneal dystrophy.

Chromosomal Location

Cytogenetic Location: 5q31.1, which is the long (q) arm of chromosome 5 at position 31.1

Molecular Location: base pairs 136,028,895 to 136,063,818 on chromosome 5 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- beta ig-h3
- BGH3_HUMAN
- BIGH3
- CDB1
- CDG2
- CDGG1
- CSD

- CSD1
- CSD2
- CSD3
- EBMD
- kerato-epithelin
- LCD1
- RGD-CAP
- RGD-containing collagen-associated protein
- transforming growth factor-beta-induced protein ig-h3
- transforming growth factor, beta-induced, 68kDa

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28TGFB1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- CORNEAL DYSTROPHY, AVELLINO TYPE
<http://omim.org/entry/607541>
- CORNEAL DYSTROPHY, EPITHELIAL BASEMENT MEMBRANE
<http://omim.org/entry/121820>
- CORNEAL DYSTROPHY, GROENOUW TYPE I
<http://omim.org/entry/121900>
- CORNEAL DYSTROPHY, LATTICE TYPE IIIA
<http://omim.org/entry/608471>
- CORNEAL DYSTROPHY, REIS-BUCKLERS TYPE
<http://omim.org/entry/608470>
- CORNEAL DYSTROPHY, THIEL-BEHNKE TYPE
<http://omim.org/entry/602082>
- TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD
<http://omim.org/entry/601692>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/TGFBIID42539ch5q31.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TGFBI%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:11771
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:7045>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7045>
- UniProt
<https://www.uniprot.org/uniprot/Q15582>

Sources for This Summary

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