KRT3 gene
keratin 3

Normal Function
The KRT3 gene provides instructions for making a protein called keratin 3. Keratins are a group of tough, fibrous proteins that form the structural framework of epithelial cells, which are cells that line the surfaces and cavities of the body. Keratin 3 is produced in a tissue on the surface of the eye called the corneal epithelium. This tissue forms the outermost layer of the cornea, which is the clear front covering of the eye. The corneal epithelium acts as a barrier to help prevent foreign materials, such as dust and bacteria, from entering the eye.

The keratin 3 protein partners with another keratin protein, keratin 12, to form molecules known as intermediate filaments. These filaments assemble into strong networks that provide strength and resilience to the corneal epithelium.

Health Conditions Related to Genetic Changes
Meesmann corneal dystrophy

At least three mutations in the KRT3 gene have been found to cause Meesmann corneal dystrophy, an eye disease characterized by the formation of tiny cysts in the corneal epithelium.

All of the identified KRT3 gene mutations associated with Meesmann corneal dystrophy change single protein building blocks (amino acids) in the keratin 3 protein. These changes occur in a region of the protein that is critical for the formation and stability of intermediate filaments. The altered keratin 3 protein interferes with the assembly of intermediate filaments, weakening the structural framework of the corneal epithelium. As a result, this outer layer of the cornea is abnormally fragile and develops the cysts that characterize Meesmann corneal dystrophy. The cysts likely contain clumps of abnormal keratin proteins and other cellular debris. When the cysts break open (rupture), they cause eye irritation, increased sensitivity to light (photophobia), and related symptoms.
**Chromosomal Location**

Cytogenetic Location: 12q13.13, which is the long (q) arm of chromosome 12 at position 13.13

Molecular Location: base pairs 52,789,685 to 52,805,735 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

**Credit:** Genome Decoration Page/NCBI

**Other Names for This Gene**

- 65 kDa cytokeratin
- CK-3
- CK3
- cytokeratin 3
- cytokeratin-3
- K2C3_HUMAN
- K3
- keratin 3, type II
- keratin, type II cytoskeletal 3
- type-II keratin Kb3

**Additional Information & Resources**

**Educational Resources**

  https://www.ncbi.nlm.nih.gov/books/NBK21560/
- National Eye Institute: Corneal Conditions
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28KRT3%5BTIAB%5D%29+OR+%28keratin+3%5BTIAB%5D%29%29+OR+%28%28CK-3%5BTIAB%5D%29%29+OR+%28cytokeratin+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- KERATIN 3, TYPE II
  http://omim.org/entry/148043

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KRT3%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3850
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P12035

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16227835
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9171831
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18806880
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