KRT12 gene

keratin 12

Normal Function

The *KRT12* gene provides instructions for making a protein called keratin 12. 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 12 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 12 protein partners with another keratin protein, keratin 3, 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 20 mutations in the *KRT12* gene have been found to cause Meesmann corneal dystrophy, an eye disease characterized by the formation of tiny cysts in the corneal epithelium.

Almost all of the *KRT12* gene mutations associated with Meesmann corneal dystrophy change single protein building blocks (amino acids) in the keratin 12 protein. These changes occur in regions of the protein that are critical for the formation and stability of intermediate filaments. The altered keratin 12 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.

Other Names for This Gene

- CK-12

- cytokeratin-12
- K12
- K1C12_HUMAN
- keratin 12, type I
- keratin, type I cytoskeletal 12
- keratin-12

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KRT12 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3859[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28KRT12%5BTIAB%5D%29+OR+%28keratin+12%5BTIAB%5D%29%29+OR+%28%28CK-12%5BTIAB%5D%29+OR+%28cytokeratin+12%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)

Catalog of Genes and Diseases from OMIM

- KERATIN 12, TYPE I; KRT12 (https://omim.org/entry/601687)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3859)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KRT12[gene])

References

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- Irvine AD, Corden LD, Swensson O, Swensson B, Moore JE, Frazer DG, Smith FJ, Knowlton RG, Christophers E, Rochels R, Uitto J, McLean WH. Mutations incornea-specific keratin K3 or K12 genes cause Meesmann's corneal dystrophy. NatGenet. 1997 Jun;16(2):184-7. doi: 10.1038/ng0697-184. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9171831)

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- Nishida K, Honma Y, Dota A, Kawasaki S, Adachi W, Nakamura T, Quantock AJ, Hosotani H, Yamamoto S, Okada M, Shimomura Y, Kinoshita S. Isolation andchromosomal localization of a cornea-specific human keratin 12 gene and detectionof four mutations in Meesmann corneal epithelial dystrophy. Am J Hum Genet. 1997Dec;61(6):1268-75. doi: 10.1086/301650. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9399908) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1716060/)

Genomic Location

The *KRT12* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

Last updated August 1, 2012