

KRT83 gene

keratin 83

Normal Function

The *KRT83* gene provides instructions for making the type II hair keratin K83 protein (K83). This protein belongs to a group of proteins known as keratins, which are tough, fibrous proteins that form the structural framework of cells that make up the hair, skin, and nails. Each keratin protein partners with another keratin protein to form molecules called intermediate filaments. These filaments assemble into strong networks that provide strength and resiliency to the tissues and protect them from being damaged by everyday physical stresses. The K83 protein is found in cells that make up the inner compartment of the hair shaft known as the cortex, and this protein helps give hair its strength and elasticity.

Health Conditions Related to Genetic Changes

Monilethrix

Mutations in the *KRT83* gene can cause monilethrix, a hair condition characterized by strands of hair with a beaded appearance and short, brittle hair that breaks easily. Mutations associated with this condition change a single protein building block (amino acid) in the K83 protein. The amino acid changes usually occur in a region of the K83 protein thought to be important in intermediate filament formation. In people with monilethrix, the cortex of the affected hair shaft appears abnormal. However, it is unclear how mutations in the *KRT83* gene are related to the abnormality in the cortex or the beaded appearance of the hair.

Erythrokeratoderma variabilis et progressiva

MedlinePlus Genetics provides information about Erythrokeratoderma variabilis et progressiva

Other Names for This Gene

- hair keratin K2.10
- hard keratin, type II, 3
- Hb-3

- HB3
- hHb3
- K83
- keratin 83, type II
- keratin, hair, basic, 3
- keratin-83
- KRT83_HUMAN
- KRTHB3
- type II hair keratin Hb3
- type-II keratin Kb23

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KRT83 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3889\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3889[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28KRT83%5BTIAB%5D%29+OR+%28hHb3%5BTIAB%5D%29+OR+%28K83%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- KERATIN 83, TYPE II; KRT83 (<https://omim.org/entry/602765>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3889>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=KRT83\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=KRT83[gene]))

References

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Genomic Location

The *KRT83* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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