KRT86 gene
keratin 86

Normal Function

The *KRT86* gene provides instructions for making the type II hair keratin K86 protein (K86). 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 K86 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

Several mutations in the *KRT86* 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 K86 protein. The amino acid changes usually occur in a region of the K86 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 *KRT86* gene are related to the abnormality in the cortex or the beaded appearance of the hair.
Chromosomal Location

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

Molecular Location: base pairs 52,270,507 to 52,309,163 on chromosome 12 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Other Names for This Gene

- hair keratin K2.11
- hard keratin, type II, 6
- HB6
- hHb6
- K86
- keratin-86
- keratin 86, type II
- keratin K-86
- keratin protein HB6
- KRT86_HUMAN
- KRTHB6
- MNX
- type II hair keratin Hb6

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Development and Structure of the Hair Follicle
  https://www.ncbi.nlm.nih.gov/books/NBK45997/#ch4829.s2
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28KRT86%5BTIAB%5D%29+OR+%28%28HB6%5BTIAB%5D%29%29+OR+%28hHb6%5BTIAB%5D%29+OR+%28K86%5BTIAB%5D%29+OR+%28KRTHB1%5BTIAB%5D%29+OR+%28KRTHB6%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+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- KERATIN 86, TYPE II
  http://omim.org/entry/601928

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KRT86.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KRT86%5Bgene%5D
- HGNC Gene Family: Keratins, type II
  https://www.genenames.org/cgi-bin/genefamilies/set/609
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnec_data.php&hgnec_id=6463
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3892
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/O43790

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12437130
- OMIM: KERATIN 86, TYPE II
  http://omim.org/entry/601928
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