

KRT81 gene

keratin 81

Normal Function

The *KRT81* gene provides instructions for making the type II hair keratin K81 protein (K81). 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 K81 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

At least two mutations in the *KRT81* gene have been identified in people with 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 K81 protein. The amino acid changes usually occur in a region of the K81 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 *KRT81* gene are related to the abnormality in the cortex or the beaded appearance of the hair.

Other Names for This Gene

- ghHb1
- hair keratin K2.9
- hard keratin, type II, 1
- Hb-1
- HB1
- hHAKB2-1
- hHb1

- keratin 81, type II
- keratin, hair, basic, 1
- keratin-81
- KRT81_HUMAN
- KRTHB1
- type II hair keratin Hb1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KRT81 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3887\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3887[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28KRT81%5BTIAB%5D%29+OR+%28%28HB1%5BTIAB%5D%29+OR+%28Hb-1%5BTIAB%5D%29+OR+%28KRTHB1%5BTIAB%5D%29+OR+%28MLN137%5BTIAB%5D%29+OR+%28hHAKB2-1%5BTIAB%5D%29+OR+%28K81%5BTIAB%5D%29+OR+%28ghHb1%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+1800+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- KERATIN 81, TYPE II; KRT81 (<https://omim.org/entry/602153>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3887>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=KRT81\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=KRT81[gene]))

References

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- Schweizer J, Bowden PE, Coulombe PA, Langbein L, Lane EB, Magin TM, Maltais L, Omary MB, Parry DA, Rogers MA, Wright MW. New consensus nomenclature for mammalian keratins. J Cell Biol. 2006 Jul 17;174(2):169-74. doi:10.1083/jcb.200603161. Epub 2006 Jul 10. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16831889>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2064177/>)

Genomic Location

The *KRT81* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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