KRT6B gene

keratin 6B

Normal Function

The *KRT6B* gene provides instructions for making a protein called keratin 6b or K6b. Keratins are a group of tough, fibrous proteins that form the structural framework of certain cells, particularly cells that make up the skin, hair, and nails. Keratin 6b is produced in the nails, the hair follicles, and the skin on the palms of the hands and soles of the feet. It is also found in the skin's sebaceous glands, which produce an oily substance called sebum.

Keratin 6b partners with a similar protein, keratin 17, to form molecules called keratin intermediate filaments. These filaments assemble into dense networks that provide strength and resilience to the skin, nails, and other tissues. Networks of keratin intermediate filaments protect these tissues from being damaged by friction and other everyday physical stresses. Keratin 6b is also among several keratins involved in wound healing.

Health Conditions Related to Genetic Changes

Pachyonychia congenita

At least four mutations in the *KRT6B* gene have been identified in people with pachyonychia congenita, a rare condition that primarily affects the nails and skin. In most cases, this condition becomes apparent within the first few months of life. These mutations either change single protein building blocks (amino acids) in keratin 6b or delete a small number of amino acids from the protein.

The *KRT6B* gene mutations responsible for pachyonychia congenita change the structure of keratin 6b, preventing it from interacting effectively with keratin 17 and interfering with the assembly of the keratin intermediate filament network. Without this network, skin cells become fragile and are easily damaged, making the skin less resistant to friction and minor trauma. Even normal activities such as walking can cause skin cells to break down, resulting in the formation of painful blisters and calluses. In the sebaceous glands, abnormal keratin filaments lead to the development of sebum-filled cysts called steatocystomas. Defective keratin 6b also disrupts the growth and function of other tissues, such as the hair follicles and nails, which explains why the signs and symptoms of pachyonychia congenita can also affect these other parts of the body.

Other Names for This Gene

- CK 6B
- CK6B
- cytokeratin 6B
- cytokeratin-6B
- K2C6B_HUMAN
- K6B
- K6b keratin
- keratin 6B, type II
- keratin, epidermal, type II, K6B
- keratin, type II cytoskeletal 6B
- KRTL1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KRT6B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3854[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28KRT6B%5BTIAB%5D%29+OR+%28keratin+6B%5BTIAB%5D%29+OR+%28cytokeratin+6B%5BTIAB%5D%29+OR+%28K6B%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- KERATIN 6B, TYPE II; KRT6B (https://omim.org/entry/148042)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3854)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KRT6B[gene])

References

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- Sharma VM, Stein SL. A novel mutation in K6b in pachyonychia congenita type 2.J Invest Dermatol. 2007 Aug;127(8):2060-2. doi: 10.1038/sj.jid.5700814. Epub 2007Apr 12. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17429440)
- Smith FJ, Jonkman MF, van Goor H, Coleman CM, Covello SP, Uitto J, McLean WH.A mutation in human keratin K6b produces a phenocopy of the K17 disorderpachyonychia congenita type 2. Hum Mol Genet. 1998 Jul;7(7):1143-8. doi: 10.1093/hmg/7.7.1143. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9618173)
- Wilson NJ, O'Toole EA, Milstone LM, Hansen CD, Shepherd AA, Al-Asadi E, Schwartz ME, McLean WH, Sprecher E, Smith FJ. The molecular genetic analysis ofthe expanding pachyonychia congenita case collection. Br J Dermatol. 2014Aug; 171(2):343-55. doi: 10.1111/bjd.12958. Epub 2014 Aug 6. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24611874) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4282083/)

Genomic Location

The *KRT6B* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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