KRT16 gene

keratin 16

Normal Function

The *KRT16* gene provides instructions for making a protein called keratin 16 or K16. 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 16 is produced in the nails, the skin on the palms of the hands and soles of the feet, and the oral mucosa that lines the inside of the mouth.

Keratin 16 partners with a similar protein, keratin 6a, 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 16 is also among several keratins involved in wound healing.

Health Conditions Related to Genetic Changes

Pachyonychia congenita

At least 19 mutations in the *KRT16* 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. Most of the *KRT16* gene mutations associated with pachyonychia congenita change single protein building blocks (amino acids) in keratin 16. A few mutations delete a small number of amino acids from the protein.

The *KRT16* gene mutations responsible for pachyonychia congenita change the structure of keratin 16, preventing it from interacting effectively with keratin 6a 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 severe, painful blisters and calluses. Additionally, fragile skin cells may abnormally produce more keratin in response to damage, which makes the skin problems worse. Defective keratin 16 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

- CK16
- cytokeratin 16
- cytokeratin-16
- K16
- K1C16_HUMAN
- K1CP
- keratin 16 (focal non-epidermolytic palmoplantar keratoderma)
- keratin 16, type I
- keratin, type I cytoskeletal 16
- keratin-16
- KRT16A
- NEPPK

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KRT16 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3868[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28KRT16%5BTIAB%5D%29+OR+%28keratin+16%5BTIAB%5D%29+OR+%28K16%5Btiab%5D++AND+keratin%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 16, TYPE I; KRT16 (https://omim.org/entry/148067)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3868)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KRT16[gene])

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Genomic Location

The *KRT16* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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