

## KRT6A gene

keratin 6A

### Normal Function

The *KRT6A* gene provides instructions for making a protein called keratin 6a or K6a. Keratins are a group of tough, fibrous proteins that form the structural framework of certain cells, particularly cells that make up the skin, hair, nails, and similar tissues. Keratin 6a 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 6a partners with a similar protein, keratin 16, 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 6a is also among several keratins involved in wound healing.

### Health Conditions Related to Genetic Changes

#### Pachyonychia congenita

About 40 mutations in the *KRT6A* 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 these mutations change single protein building blocks (amino acids) in keratin 6a. A few mutations add or delete a small number of amino acids.

The *KRT6A* gene mutations responsible for pachyonychia congenita change the structure of keratin 6a, preventing it from interacting effectively with keratin 16 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 6a 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

- 56 cytoskeletal type II keratin
- CK 6A
- CK6A
- CK6C
- CK6D
- cytokeratin 6A
- cytokeratin-6A
- K2C6A\_HUMAN
- K6A
- K6C
- K6D
- keratin 6A, type II
- keratin, epidermal type II, K6A

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of KRT6A ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3853\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3853[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28KRT6A%5BTIAB%5D%29+OR+%28keratin+6A%5BTIAB%5D%29%29+OR+%28K6A%5BTIAB%5D%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 6A, TYPE II; KRT6A (<https://omim.org/entry/148041>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3853>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=KRT6A\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=KRT6A[gene]))

## References

- Bowden PE, Haley JL, Kansky A, Rothnagel JA, Jones DO, Turner RJ. Mutation of a type II keratin gene (K6a) in pachyonychia congenita. *Nat Genet.* 1995 Jul;10(3):363-5. doi: 10.1038/ng0795-363. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/7545493>)
- Liao H, Sayers JM, Wilson NJ, Irvine AD, Mellerio JE, Baselga E, Bayliss SJ, Uliana V, Fimiani M, Lane EB, McLean WH, Leachman SA, Smith FJ. A spectrum of mutations in keratins K6a, K16 and K17 causing pachyonychia congenita. *J Dermatol Sci.* 2007 Dec;48(3):199-205. doi: 10.1016/j.jdermsci.2007.07.003. Epub 2007 Aug 24. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17719747>)
- McLean WH, Hansen CD, Eliason MJ, Smith FJ. The phenotypic and molecular genetic features of pachyonychia congenita. *J Invest Dermatol.* 2011 May;131(5):1015-7. doi: 10.1038/jid.2011.59. Epub 2011 Mar 24. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21430705>)
- Spaunhurst KM, Hogendorf AM, Smith FJ, Lingala B, Schwartz ME, Cywinska-Bernas A, Zeman KJ, Tang JY. Pachyonychia congenita patients with mutations in KRT6A have more extensive disease compared with patients who have mutations in KRT16. *Br J Dermatol.* 2012 Apr;166(4):875-8. doi: 10.1111/j.1365-2133.2011.10745.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22098151>)
- Terrinoni A, Smith FJ, Didona B, Canzona F, Paradisi M, Huber M, Hohl D, David A, Verloes A, Leigh IM, Munro CS, Melino G, McLean WH. Novel and recurrent mutations in the genes encoding keratins K6a, K16 and K17 in 13 cases of pachyonychia congenita. *J Invest Dermatol.* 2001 Dec;117(6):1391-6. doi: 10.1046/j.0022-202x.2001.01565.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11886499>)
- Wilson NJ, Toole EA, Milstone LM, Hansen CD, Shepherd AA, Al-Asadi E, Schwartz ME, McLean WH, Sprecher E, Smith FJ. The molecular genetic analysis of the expanding pachyonychia congenita case collection. *Br J Dermatol.* 2014 Aug;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 *KRT6A* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

**Last updated December 1, 2015**