



## KRT6C gene

keratin 6C

### Normal Function

The *KRT6C* gene provides instructions for making a protein called keratin 6c or K6c. 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 6c is found in the skin, although it is unknown which other tissues may produce this protein.

Keratin 6c is a component of 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.

### Health Conditions Related to Genetic Changes

#### Pachyonychia congenita

At least four mutations in the *KRT6C* gene have been found to cause 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.

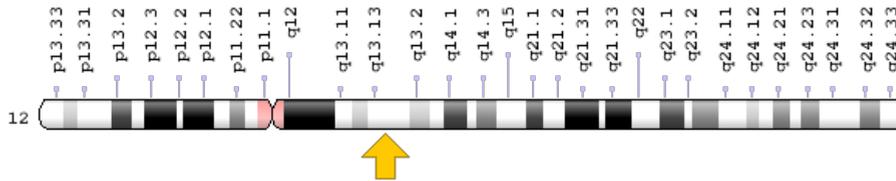
One of the mutations associated with pachyonychia congenita changes a single protein building block (amino acid) in the keratin 6c protein. Specifically, this mutation replaces the amino acid glutamic acid with the amino acid lysine at protein position 472 (written as Glu472Lys or E472K). The other *KRT6C* gene mutations delete one or more amino acids from the keratin 6c protein.

All of the known *KRT6C* gene mutations alter the structure of keratin 6c and interfere 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 6c 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.

## Chromosomal Location

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

Molecular Location: base pairs 52,468,516 to 52,473,785 on chromosome 12 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CK-6C
- CK-6E
- cytokeratin-6C
- cytokeratin-6E
- K2C6C\_HUMAN
- K6C
- K6E
- keratin-6C
- keratin 6C, type II
- keratin 6E
- keratin K6h
- keratin, type II cytoskeletal 6C
- KRT6E
- type-II keratin Kb12

## Additional Information & Resources

### Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Intermediate Filaments  
<https://www.ncbi.nlm.nih.gov/books/NBK9834/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28KRT6C%5BTIAB%5D%29+OR+%28keratin+6C%5BTIAB%5D%29%29+OR+%28K6C%5BTIAB%5D%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 6C, TYPE II  
<http://omim.org/entry/612315>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=KRT6C%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:20406](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:20406)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:286887>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/286887>
- UniProt  
<https://www.uniprot.org/uniprot/P48668>

### **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/KRT6C>

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