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

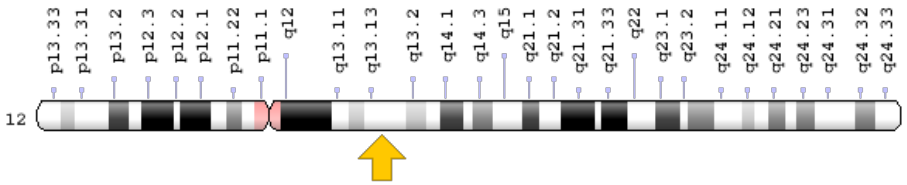
About 40 mutations in the *KRT6A* gene have been identified in people with pachyonychia congenita, a rare condition that primarily affects the nails and skin. In most cases, this condition becomes apparent within the first few months of life. Most of these mutations change single protein building blocks (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.

Chromosomal Location

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

Molecular Location: base pairs 52,487,176 to 52,493,257 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Keratin Filaments in Epithelial Cells (image)
  https://www.ncbi.nlm.nih.gov/books/NBK26862/figure/A2988/
- The Cell: A Molecular Approach (second edition, 2000): Intermediate Filaments
  https://www.ncbi.nlm.nih.gov/books/NBK9834/

Clinical Information from GeneReviews

- Pachyonychia Congenita
  https://www.ncbi.nlm.nih.gov/books/NBK1280

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

Catalog of Genes and Diseases from OMIM

- KERATIN 6A, TYPE II
  http://omim.org/entry/148041

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KRT6A.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KRT6A%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6443
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3853
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3853
- UniProt
  https://www.uniprot.org/uniprot/P02538

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