KRT4 gene

keratin 4

Normal Function

The *KRT4* gene provides instructions for making a protein called keratin 4. Keratins are a group of tough, fibrous proteins that form the structural framework of epithelial cells, which are cells that line the surfaces and cavities of the body. Keratin 4 is found in the moist lining (mucosae) of the mouth, nose, esophagus, genitals, and anus.

Keratin 4 partners with a similar protein, keratin 13 (produced from the *KRT13* gene), to form molecules known as intermediate filaments. These filaments assemble into strong networks that provide strength and resilience to the different mucosae. Networks of intermediate filaments protect the mucosae from being damaged by friction or other everyday physical stresses.

Health Conditions Related to Genetic Changes

White sponge nevus

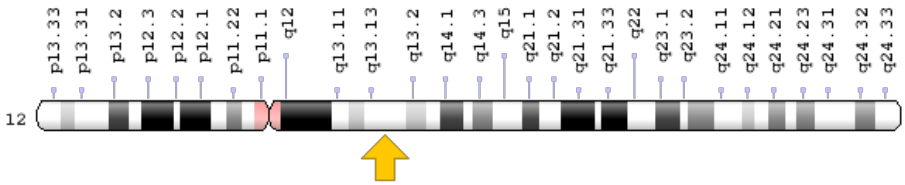
At least six mutations in the *KRT4* gene have been found to cause white sponge nevus, a condition that results in the formation of white patches of tissue called nevi (singular: nevus) that appear as thickened, velvety, sponge-like tissue. These nevi most often occur on the mouth (oral) mucosa (plural: mucosae). Rarely, white sponge nevus occurs on the mucosae of the nose, esophagus, genitals, or anus.

The *KRT4* gene mutations that cause white sponge nevus disrupt the structure of keratin 4. As a result, keratin 4 does not fit together properly with keratin 13, leading to the formation of irregular intermediate filaments that are easily damaged with little friction or trauma. Fragile intermediate filaments in the oral mucosa might be damaged when eating or brushing one's teeth. Damage to intermediate filaments leads to inflammation and promotes the abnormal growth and division (proliferation) of epithelial cells, causing the mucosae to thicken and resulting in white sponge nevus.

Chromosomal Location

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

Molecular Location: base pairs 52,806,543 to 52,814,116 on chromosome 12 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CK-4
- CK4
- CYK4
- cytokeratin 4
- K4
- keratin 4, type II
- keratin, type II cytoskeletal 4
- type-II keratin Kb4

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Keratin Intermediate Filaments and Diseases of the Skin
  https://www.ncbi.nlm.nih.gov/books/NBK6247/
- The Cell: A Molecular Approach (2nd 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%28KRT4%5BTIAB%5D%29+OR+%28keratin+4%5BTIAB%5D%29%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 4, TYPE II
  http://omim.org/entry/123940

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KRT4.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KRT4%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6441
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3851
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3851

Sources for This Summary

- OMIM: KERATIN 4, TYPE II
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- Shimizu A, Yokoyama Y, Shimomura Y, Ishikawa O. White sponge nevus caused by a missense mutation in the keratin 4 gene. Eur J Dermatol. 2012 Jul-Aug;22(4):571-2. doi: 10.1684/ejd.2012.1765.
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- Zhang JM, Yang ZW, Chen RY, Gao P, Zhang YR, Zhang LF. Two new mutations in the keratin 4 gene causing oral white sponge nevus in Chinese family. Oral Dis. 2009 Jan;15(1):100-5. doi: 10.1111/j.1601-0825.2008.01498.x. Epub 2008 Oct 25.
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