KRT13 gene

keratin 13

Normal Function

The *KRT13* gene provides instructions for making a protein called keratin 13. 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 13 is found in the moist lining (mucosae) of the mouth, nose, esophagus, genitals, and anus.

Keratin 13 partners with a similar protein, keratin 4 (produced from the *KRT4* 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 five *KRT13* gene mutations 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 *KRT13* gene mutations that cause white sponge nevus disrupt the structure of keratin 13. As a result, keratin 13 does not fit together properly with keratin 4, 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.

Other Names for This Gene

- CK-13
- CK13
- cytokeratin 13

- cytokeratin-13
- K13
- K1C13_HUMAN
- keratin 13, type I
- keratin, type I cytoskeletal 13
- keratin-13

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KRT13 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3860[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28KRT13%5BTIAB%5D%29+OR+%28keratin+13%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 13, TYPE I; KRT13 (https://omim.org/entry/148065)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3860)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KRT13[gene])

References

- Nishizawa A, Nakajima R, Nakano H, Sawamura D, Takayama K, Satoh T, YokozekiH. A de novo missense mutation in the keratin 13 gene in oral white spongenaevus. Br J Dermatol. 2008 Sep;159(4):974-5. doi:10.1111/j.1365-2133.2008.08716.x. Epub 2008 Jul 4. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18616775)
- Rugg E, Magee G, Wilson N, Brandrup F, Hamburger J, Lane E. Identification oftwo novel mutations in keratin 13 as the cause of white sponge naevus. Oral Dis.1999 Oct;5(4):321-4. doi: 10.1111/j.1601-0825.1999.tb00097.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10561721)
- Shibuya Y, Zhang J, Yokoo S, Umeda M, Komori T. Constitutional mutation

ofkeratin 13 gene in familial white sponge nevus. Oral Surg Oral Med Oral PatholOral Radiol Endod. 2003 Nov;96(5):561-5. doi: 10.1016/s1079-2104(03)00372-x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14600690)

Genomic Location

The *KRT13* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

Last updated February 1, 2014