Steatocystoma multiplex

Steatocystoma multiplex is a skin disorder characterized by the development of multiple noncancerous (benign) cysts known as steatocystomas. These growths begin in the skin’s sebaceous glands, which normally produce an oily substance called sebum that lubricates the skin and hair. Steatocystomas are filled with sebum.

In affected individuals, steatocystomas typically first appear during adolescence and are found most often on the torso, neck, upper arms, and upper legs. These cysts are usually the only sign of the condition. However, some affected individuals also have mild abnormalities involving the teeth or the fingernails and toenails.

Frequency

Although the prevalence of steatocystoma multiplex is unknown, it appears to be rare.

Causes

Steatocystoma multiplex can be caused by mutations in the KRT17 gene. This gene provides instructions for making a protein called keratin 17, which is produced in the nails, the hair follicles, and the skin on the palms of the hands and soles of the feet. It is also found in the skin’s sebaceous glands. Keratin 17 partners with a similar protein called keratin 6b to form networks that provide strength and resilience to the skin, nails, and other tissues.

The KRT17 gene mutations that cause steatocystoma multiplex alter the structure of keratin 17, preventing it from forming strong, stable networks within cells. The defective keratin network disrupts the growth and function of cells in the skin and nails, including cells that make up the sebaceous glands. These abnormalities lead to the growth of sebum-containing cysts in people with steatocystoma multiplex. However, it is unclear why steatocystomas are typically the only feature of this disorder.

Many researchers believe that steatocystoma multiplex is a variant form of a disorder called pachyonychia congenita, which can also result from mutations in the KRT17 gene. Like steatocystoma multiplex, pachyonychia congenita involves the growth of steatocystomas. Pachyonychia congenita is also associated with more severe nail abnormalities and a painful skin condition called palmoplantar keratoderma, which are not usually found in people with steatocystoma multiplex.

In some cases, people with steatocystoma multiplex do not have an identified mutation in the KRT17 gene. The cause of the condition in these individuals is unknown.
Inheritance Pattern

When steatocystoma multiplex is caused by mutations in the KRT17 gene, it is inherited in an autosomal dominant pattern. Autosomal dominant inheritance means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person inherits the condition from an affected parent.

In people with steatocystoma multiplex who do not have identified KRT17 gene mutations, there is usually no family history of the disorder.

Other Names for This Condition

• multiple sebaceous cysts
• multiplex steatocystoma
• sebocystomatosis

Diagnosis & Management

Genetic Testing Information

• What is genetic testing? /primer/testing/genetictesting
• Genetic Testing Registry: Steatocystoma multiplex

Additional Information & Resources

Health Information from MedlinePlus

• Encyclopedia: Hair Follicle Sebaceous Gland
  https://medlineplus.gov/ency/imagepages/19666.htm
• Health Topic: Skin Conditions
  https://medlineplus.gov/skinconditions.html

Genetic and Rare Diseases Information Center

• Steatocystoma multiplex
  https://rarediseases.info.nih.gov/diseases/5003/steatocystoma-multiplex

Educational Resources

• MalaCards: steatocystoma multiplex
  https://www.malacards.org/card/steatocystoma_multiplex
• Orphanet: Steatocystoma multiplex-natal teeth syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3184
Patient Support and Advocacy Resources

- Ectodermal Dysplasia Society
  https://edsociety.co.uk/

- National Foundation for Ectodermal Dysplasias
  https://www.nfed.org/

- Pachyonychia Congenita Project
  http://www.pachyonychia.org/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28steatocystoma+multiplex%5BTIAB%5D%29+OR+%28sebocystomatosis%5BTIAB%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

- STEATOCYSTOMA MULTIPLEX
  http://omim.org/entry/184500

Sources for This Summary


Reprinted from Genetics Home Reference:

Reviewed: September 2016
Published: October 9, 2018