Scalp-ear-nipple syndrome

Scalp-ear-nipple syndrome, as its name suggests, is a condition characterized by abnormalities of the scalp, ears, and nipples. Less frequently, affected individuals have problems affecting other parts of the body. The features of this disorder can vary even within the same family.

Babies with scalp-ear-nipple syndrome are born with a condition called aplasia cutis congenita, which involves patchy abnormal areas (lesions) on the scalp. These lesions are firm, raised, hairless nodules that resemble open wounds or ulcers at birth, but that heal during childhood.

The external ears of people with scalp-ear-nipple syndrome may be small, cup-shaped, folded over, or otherwise mildly misshapen. Hearing is generally normal. Affected individuals also have nipples that are underdeveloped (hypothelia) or absent (athelia). In some cases the underlying breast tissue is absent as well (amastia).

Other features that can occur in this disorder include malformed and brittle fingernails and toenails (nail dystrophy), dental abnormalities including widely-spaced or missing teeth, fusion of the skin between some of the fingers and toes (cutaneous syndactyly), and kidney defects such as underdevelopment (hypoplasia) of one or both kidneys. Unusual facial features, including narrowed openings of the eyes (narrowed palpebral fissures), an increased distance between the inner corners of the eyes (telecanthus), a flat bridge of the nose, and nostrils that open to the front rather than downward (anteverted nares), can also occur in this disorder.

Frequency

The prevalence of scalp-ear-nipple syndrome is unknown. Only a small number of affected individuals have been described in the medical literature.

Causes

Scalp-ear-nipple syndrome is caused by mutations in the \textit{KCTD1} gene. This gene provides instructions for making a protein that acts as a transcriptional repressor, which means that it turns off (represses) the activity of certain genes when they are not needed. Specifically, the KCTD1 protein is thought to control (regulate) the activity of genes involved in the development of an embryonic cell layer called the ectoderm. Within the developing embryo, the ectoderm gives rise to several body tissues including the skin, hair, nails, and teeth.

The mutations in the \textit{KCTD1} gene that cause scalp-ear-nipple syndrome impair the transcriptional repressor function of the KCTD1 protein. Impairment of this function results in abnormal regulation of genes involved in ectodermal development. The
altered gene activity disrupts normal development of the tissues that arise from the ectoderm (ectodermal dysplasia) and leads to the signs and symptoms of scalp-ear-nipple syndrome.

Inheritance Pattern
Scalp-ear-nipple syndrome is considered to be an autosomal dominant condition, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most cases of this condition result from new mutations in the gene and occur in people with no history of the disorder in their family. In other cases, an affected person inherits the mutation from one affected parent.

Other Names for This Condition
- Finlay-Marks syndrome
- hereditary syndrome of lumpy scalp, odd ears, and rudimentary nipples
- SEN syndrome
- SENS

Diagnosis & Management

Genetic Testing Information
- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Scalp ear nipple syndrome

Additional Information & Resources

Health Information from MedlinePlus
- Encyclopedia: Ectodermal dysplasia
  https://medlineplus.gov/ency/article/001469.htm
- Health Topic: Skin Conditions
  https://medlineplus.gov/skinconditions.html

Genetic and Rare Diseases Information Center
- Scalp ear nipple syndrome
Educational Resources

• MalaCards: scalp-ear-nipple syndrome
  https://www.malacards.org/card/scalp_ear_nipple_syndrome_2

• Orphanet: Scalp-ear-nipple syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2036

Patient Support and Advocacy Resources

• American Skin Association
  http://www.americanskin.org/

• Children's Skin Disease Foundation
  https://www.csdf.org/

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28scalp-ear-nipple%5BTIAB%5D%29+OR+%28Finlay-Marks%5BTIAB%5D%29+OR+%28lumpy+scalp,+odd+ears+and+rudimentary+nipples%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmhr%5D

Catalog of Genes and Diseases from OMIM

• SCALP-EAR-NIPPLE SYNDROME
  http://omim.org/entry/181270

Medical Genetics Database from MedGen

• Scalp ear nipple syndrome

Sources for This Summary


