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My46 Trait Profile

Scalp-ear-nipple syndrome

Other Names: Finlay-Marks syndrome

Scalp-ear-nipple syndrome is a rare genetic condition that causes anomalies in the scalp, ears, and nipples. It is caused by mutations in the KCTD1 gene that plays an important role in the regulation of other genes in the body.

Characteristics of Scalp-ear-nipple syndrome

Scalp-ear-nipple syndrome is a rare genetic condition that causes congenital (i.e., present at birth) anomalies involving the scalp, ears, and nipples. Scalp-ear-nipple syndrome causes an absence of skin (aplasia cutis congenita) of the scalp, and minor malformation of the external ears. Breast abnormalities range from missing a nipple (hypothelia), absent nipples (athelia), to no breast tissue (amastia). Scalp-ear-nipple syndrome may also cause dental anomalies, kidney abnormalities, thin or missing hair, webbed fingers or toes (cutaneous syndactyly), and brittle nails. The signs and symptoms of scalp-ear-nipple syndrome are highly variable within families as well as between other unrelated, affected individuals.

Diagnosis/Testing

Most individuals with scalp-ear-nipple syndrome have a change or mutation in a gene called KCTD1. This gene plays an important role in the regulation of other genes in the body.

Management/Surveillance

Management of scalp-ear-nipple syndrome is based on signs and symptoms. Medical treatment of cutis aplasia may involve the use of topical ointments to prevent the remaining thin membrane from drying out. The damaged area usually heals on its own.

Mode of inheritance

Scalp-ear-nipple syndrome is inherited in an autosomal dominant pattern. This means inheriting one KCTD1 mutation is enough for an individual to be affected and show signs of scalp-ear-nipple syndrome. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with scalp-ear-nipple syndrome has a parent affected with the condition. If a parent also has scalp-ear-nipple syndrome, the risk of having a child with the condition is 50% with each pregnancy. If a parent does not have scalp-ear-nipple syndrome, the risk of other siblings being affected is very low.

Special considerations

None

Resources

NIH Office of Rare Diseases Research: Scalp ear nipple syndrome

http://rarediseases.info.nih.gov/gard/159/scalp-ear-nipple-syndrome/resources/5

References

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