Acral peeling skin syndrome

Acral peeling skin syndrome is a skin disorder characterized by painless peeling of the top layer of skin. The term "acral" refers to the fact that the skin peeling in this condition is most apparent on the hands and feet. Occasionally, peeling also occurs on the arms and legs. The peeling is usually evident from birth, although the condition can also begin in childhood or later in life. Skin peeling is made worse by exposure to heat, humidity and other forms of moisture, and friction. The underlying skin may be temporarily red and itchy, but it typically heals without scarring. Acral peeling skin syndrome is not associated with any other health problems.

Frequency

Acral peeling skin syndrome is a rare condition, with several dozen cases reported in the medical literature. However, because its signs and symptoms tend to be mild and similar to those of other skin disorders, the condition is likely underdiagnosed.

Causes

Acral peeling skin syndrome is caused by mutations in the *TGM5* gene. This gene provides instructions for making an enzyme called transglutaminase 5, which is a component of the outer layer of skin (the epidermis). Transglutaminase 5 plays a critical role in the formation of a structure called the cornified cell envelope, which surrounds epidermal cells and helps the skin form a protective barrier between the body and its environment.

TGM5 gene mutations reduce the production of transglutaminase 5 or prevent cells from making any of this protein. A shortage of transglutaminase 5 weakens the cornified cell envelope, which allows the outermost cells of the epidermis to separate easily from the underlying skin and peel off. This peeling is most noticeable on the hands and feet probably because those areas tend to be heavily exposed to moisture and friction.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- APSS
- peeling skin syndrome, acral type

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Peeling skin syndrome, acral type
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853354/

Other Diagnosis and Management Resources

- Birmingham Children's Hospital, National Health Service (UK)
  https://www.debra.org.uk/downloads/community-support/acral-peeling-skin-syndrome.pdf

Additional Information & Resources

Health Information from MedlinePlus

- Health Topic: Skin Conditions
  https://medlineplus.gov/skinconditions.html

Genetic and Rare Diseases Information Center

- Acral peeling skin syndrome
  https://rarediseases.info.nih.gov/diseases/12863/acral-peeling-skin-syndrome

Educational Resources

- Birmingham Children's Hospital, National Health Service (UK)
  https://www.debra.org.uk/downloads/community-support/acral-peeling-skin-syndrome.pdf
- Orphanet: Acral peeling skin syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=263534

Patient Support and Advocacy Resources

- Foundation for Ichthyosis and Related Skin Types (FIRST)
  http://www.firstskinfoundation.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/peeling-skin-syndrome/
- Resource List from the University of Kansas Medical Center: Dermatology and Genetics
  http://www.kumc.edu/gec/support/derm.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28peeling+skin+syndrome%5BTIAB%5D%29+AND+%28acral%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- PEELING SKIN SYNDROME 2
  http://omim.org/entry/609796

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