Peeling skin syndrome 2

Description

Peeling skin syndrome 2 is a skin disorder characterized by painless peeling of the top layer of skin. In this form of peeling skin syndrome, the peeling is most apparent on the hands and feet. Occasionally, peeling also occurs on the arms and legs. The peeling usually starts soon after 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. Peeling skin syndrome 2 is not associated with any other health problems.

Frequency

Peeling skin syndrome 2 is a rare condition, with a few 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

Peeling skin syndrome 2 is caused by variants (also called mutations) in the *TGM5* gene. This gene provides instructions for making an enzyme called transglutaminase 5, which is part 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 variants reduce the activity of transglutaminase 5 or prevent cells from making any of this enzyme. 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. The peeling may be most noticeable on the hands and feet because those areas are more often exposed to moisture and friction.

Learn more about the gene associated with Peeling skin syndrome 2

- TGM5

Inheritance

Peeling skin syndrome 2 is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Acral peeling skin syndrome
- APSS
- Peeling skin syndrome, acral type

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Acral peeling skin syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853354/)

Genetic and Rare Diseases Information Center

- Acral peeling skin syndrome (https://rarediseases.info.nih.gov/diseases/12863/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- PEELING SKIN SYNDROME 2; PSS2 (https://omim.org/entry/609796)

Scientific Articles on PubMed

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

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