Acral peeling skin syndrome

Description

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.

Learn more about the gene associated with Acral peeling skin syndrome

- TGM5

Inheritance

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

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/acral-peeling-skin-syndrome)

Patient Support and Advocacy Resources

- Disease InfoSearch (https://www.diseaseinfosearch.org/)
- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- PEELING SKIN SYNDROME 2 (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)

References

- Cassidy AJ, van Steensel MA, Steijlen PM, van Geel M, van der Velden J, MorleySM, Terrinoni A, Melino G, Candi E, McLean WH. A homozygous missense

mutation inTGM5 abolishes epidermal transglutaminase 5 activity and causes acral peelingskin syndrome. Am J Hum Genet. 2005 Dec;77(6):909-17. doi: 10.1086/497707. Epub2005 Oct 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16380904) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1285176/)

- Kiritsi D, Cosgarea I, Franzke CW, Schumann H, Oji V, Kohlhase J,Bruckner-Tuderman L, Has C. Acral peeling skin syndrome with TGM5 gene mutationsmay resemble epidermolysis bullosa simplex in young individuals. J InvestDermatol. 2010 Jun;130(6):1741-6. doi: 10.1038/jid.2010.23. Epub 2010 Feb 18. Noabstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20164844)
- Pavlovic S, Krunic AL, Bulj TK, Medenica MM, Fong K, Arita K, McGrath JA.Acral peeling skin syndrome: a clinically and genetically heterogeneous disorder.Pediatr Dermatol. 2012 May-Jun;29(3):258-63. doi:10.1111/j.1525-1470.2011.01563.x. Epub 2011 Nov 8. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22066523)
- Pigors M, Kiritsi D, Cobzaru C, Schwieger-Briel A, Suarez J, Faletra F, Aho H, Makela L, Kern JS, Bruckner-Tuderman L, Has C. TGM5 mutations impact epidermaldifferentiation in acral peeling skin syndrome. J Invest Dermatol. 2012Oct;132(10):2422-2429. doi: 10.1038/jid.2012.166. Epub 2012 May 24. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22622422)
- Szczecinska W, Nesteruk D, Wertheim-Tysarowska K, Greenblatt DT, Baty D, Browne F, Liu L, Ozoemena L, Terron-Kwiatkowski A, McGrath JA, Mellerio JE, Morton J, Wozniak K, Kowalewski C, Has C, Moss C. Under-recognition of acralpeeling skin syndrome: 59 new cases with 15 novel mutations. Br J Dermatol. 2014Nov;171(5):1206-10. doi: 10.1111/bjd.12964. Epub 2014 Oct 20. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24628291)

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