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Review Article

Medical & Clinical Research

The Role of Genetics Mutation in Gene Tgm5 in Induce Acral Peeling Skin Syndrome

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Abstract

APS syndrome is a skin-related genetic disorder characterized by painless skin lesions from the upper layer of the skin. In addition to the above, sometimes peeling of the skin in the arms and legs also occurs. Skin peeling usually appears at birth, but can begin in childhood or later in life. APS syndrome is caused by the mutation of the TGM5 gene, which is based on the long arm of chromosome 15 as 15q15.2.

Keywords: Acral Peeling Skin syndrome, TGM5 gene, Skin disorder.

General of the Acarel Peeling Skin Syndrome (APS)

APS syndrome is a skin-related genetic disorder characterized by painless skin lesions from the upper layer of the skin. The term Acral points out that the peeling of the skin in the syndrome takes place more on the skin of the hands and feet [1].

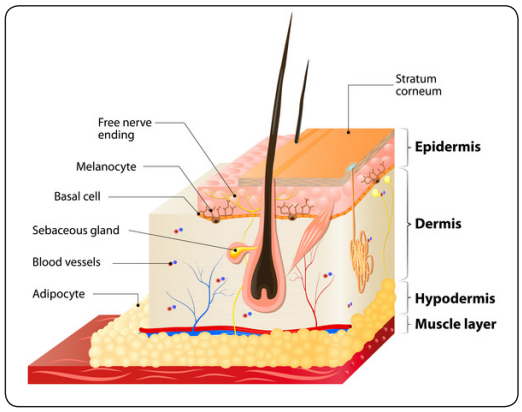

Figure 1: Schematic of the structure of skin layers

Clinical Signs and Symptoms of Acarel Peeling Skin Syndrome (APS)

In addition to the above, sometimes peeling of the skin in the arms and legs also occurs. Skin peeling usually appears at birth, but can begin in childhood or later in life. Skin peeling worse when exposed to heat, moisture and friction in APS syndrome patients. The undercoat may be temporarily red and itchy, but usually heals without wounds. It is worth noting that APS syndrome, with the exception of the above, is not associated with any other health and health problems [2].

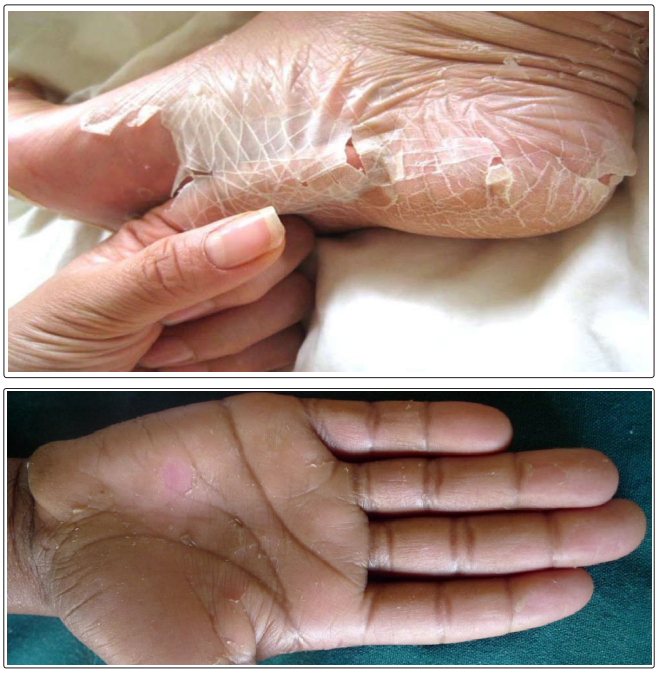

Figure 2: Images of the hands and feet of the man with APS syndrome, with dysmotility in the palms of the hands and feet

Etiology of Acral Peeling Skin Syndrome (APS)

APS syndrome is caused by the mutation of the TGM5 gene, which is based on the long arm of chromosome 15 as 15q15.2. The gene provides instructions for the synthesis of an enzyme called transglutaminase 5, which is the main component of the outer skin layer (epidermis). This enzyme plays an important role in the formation of a structure called cellular coating that surrounds the epidermal cells and helps the skin to create a barrier between body and environment [3].

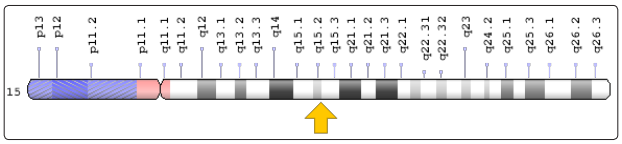

Figure 3: Schematic view of Chromosome No. 15 in which the TGM5 gene is located in the long arm of this chromosome 15q15.2

TGM5 gene mutations reduce the production of transglutaminase 5 or prevent the production of cells from this enzyme. The lack of transglutaminase 5 reduces the thickness of the cells, and allows the epidermal cells to be easily removed from the underlying skin. This layer is noticeable on the hands and feet, as they are heavily exposed to moisture and friction [3].

The APS syndrome follows an autosomal recessive hereditary pattern. Therefore, in order to produce this syndrome, two copies of the mutated gene of TGM5 (one parent and one of the mother) are needed, and the chance of having a child with autosomal recessive syndrome is 25% for each pregnancy [4].

Frequency of Acral Peeling Skin syndrome

APS syndrome is a rare skin disorder that has been reported in only a few cases in the medical literature ever since [4].

Diagnosis of Acral Peeling Skin syndrome

The APS syndrome is diagnosed based on the clinical and physical findings of the patients and some pathological and dermatological tests. The most accurate method for detecting this syndrome is the molecular genetic testing of the TGM5 gene to investigate the presence of possible mutations [5].

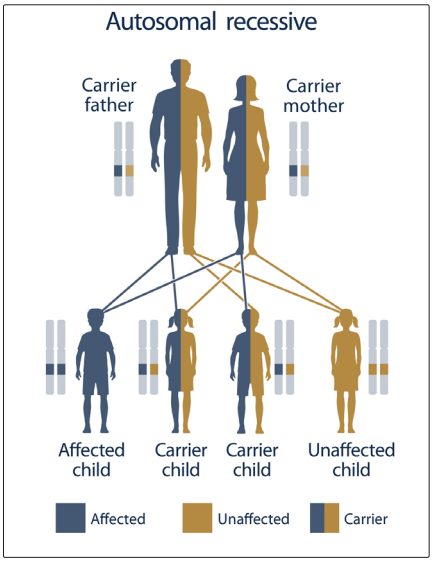

Figure 4: schematic view of an autosomal recessive hereditary pattern that follows the APS syndrome

Treatment pathways for Acral Peeling Skin syndrome

The strategy of treatment and management of APS syndrome is symptomatic and supportive. Treatment may be done by a team of experts, including dermatologist and skin surgeon. There is no reliable treatment for this syndrome, and all clinical measures are designed to reduce the suffering of the sufferers. Genetic counseling is also needed for all parents who want a healthy baby [5].

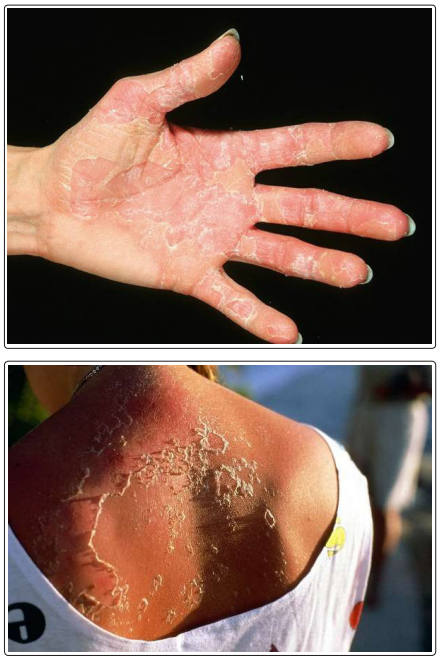

Figure 5: Other pictures of the skin of APS syndrome patients with dysplasia

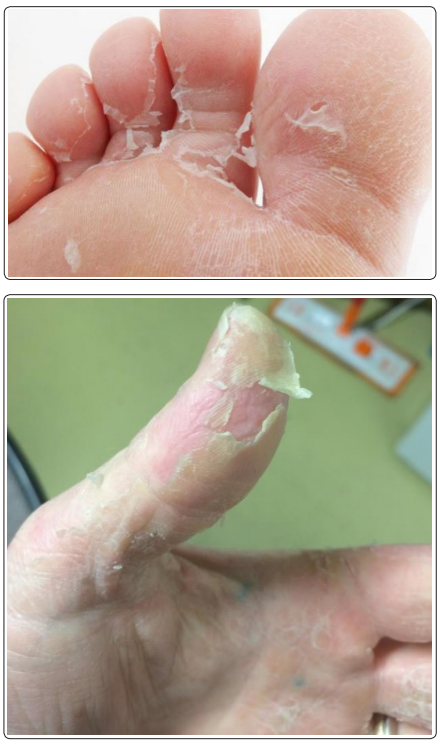

Figure 6: A Overview of Dysplasia Disorder in APS Syndrome

Discussion and conclusion

APS syndrome is a skin-related genetic disorder characterized by painless skin lesions from the upper layer of the skin. In addition to the above, sometimes peeling of the skin in the arms and legs also occurs. Skin peeling usually appears at birth, but can begin in childhood or later in life. APS syndrome is caused by the mutation of the TGM5 gene, which is based on the long arm of chromosome 15 as 15q15.2. There is no reliable treatment for this syndrome,

and all clinical measures are designed to reduce the suffering of the sufferers.

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