

## Peeling skin syndrome in a 3 year old female child

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**Abstract:** We report a case of 3 year old Hindu female child who presented to skin OPD with complaint of spontaneous peeling of skin from complete body which was insidious in onset and progressive in nature, started since 10<sup>th</sup> day of birth mainly involving frictional sites. History of similar illness is found in father of the child.

**Key words:** Peeling skin syndrome, TGM5 gene, Acral skin peeling syndrome

**Key message:** Peeling skin syndrome is an autosomal recessive dermatosis due to defective TGM5 gene, characterized by shedding of skin throughout life.

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### I. Introduction

Peeling skin syndrome is a rare genodermatosis with autosomal recessive inheritance characterized by the continuous shedding of the outer layers of the epidermis from birth and throughout life.<sup>[1]</sup> Clinically characterised by painless, continual and spontaneous skin peeling since birth associated with itching, redness and formation of blisters.

Generalized PSS has been subclassified into a noninflammatory (type A) manifest white scaling, with painless and easy removal of the skin, and an inflammatory (type B)<sup>[2]</sup> in which generalized peeling of skin is associated with pruritus and atopy.

Another variant where the skin peeling is predominantly present at the hands and feet is known as Acral peeling skin syndrome.

The main pathogenesis being the autosomal recessive inheritance of the defective TGM5 gene coding for transglutaminase enzyme which helps forming the corneodesmosomes. Deficient 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.

Histologically the level of blistering is characterized by separation of the epidermis between the stratum corneum and the stratum granulosum.<sup>[2]</sup>

### II. Case Report

A 3-year old female child presented to skin OPD accompanied with the mother complaining of generalised peeling of skin from complete body since birth starting as early as 10th day of birth beginning from the diaper area progressively involving frictional sites as the child grew.

Multiple ill-defined areas of post inflammatory hyperpigmentation can be observed at the wrists, dorsum of feet, and trunk (img. 1). Multiple erosions of varying size present over trunk and buttocks (img. 2). Areas of dry, asteototic skin with fine scaling can be observed over bilateral cheeks, forearms (img. 3).

There is no history of seasonal predilection, although improvement in the general condition and decreased frequency of episodes with age are noticed.

History of similar illness in father is present.

Skin biopsy is sent from the latest vesicle/ erosion where the histology shows the level of blistering is high in the epidermis, at the junction of the stratum granulosum and the stratum corneum.

History, Clinical and histopathological examination confirm the diagnosis of peeling skin syndrome.

### III. Discussion

Peeling skin syndrome is characterized by generalized, continuous shedding of the outer layers of the epidermis which may be pruritic or nonpruritic and sometimes accompanied by erythema or vesiculation. The skin involvement is usually general, but in some patients the scalp, face, palms, and soles may be unaffected. Seasonal changes have been reported. Two main subtypes include non-inflammatory (type A), a continuous nonerythematous exfoliation, which is usually congenital or appears during childhood. Inflammatory (type B)

which is associated with generalized erythema, pruritus and atopy - an ichthyosiform erythroderma characterized by lifelong patchy peeling of the entire skin with onset at birth or shortly after.<sup>[3]</sup>

Genetic Heterogeneity of Peeling Skin Syndrome-

Peeling skin syndrome-2 (PSS2) an acral form of the disorder that mainly involves palmar and plantar skin, is caused by mutation in the TGM5 gene on chromosome 15q15.

Peeling skin syndrome-3 (PSS3) is caused by mutation in the CHST8 gene on chromosome 19q13.

Peeling skin syndrome-4 (PSS4) is caused by mutation in the CSTA gene on chromosome 3q21.

Peeling skin syndrome-5 (PSS5) is caused by mutation in the SERPINB8 gene on chromosome 18q22.

PSS6 is caused by mutation in the FLG2 gene on chromosome 1q21<sup>[2]</sup>.

The primary deficient gene is TGM5 codes for Transglutaminases (TGs) which are involved in protein cross-linking by catalyzing the formation of gamma-glutamyl-lysine iso-dipeptide bonds between adjacent polypeptides.<sup>[4]</sup> This process is particularly important in the terminal differentiation of the epidermis, where TGs heavily cross-link keratins and a range of differentiation-specific structural proteins, such as involucrin, loricrin, filaggrin, and small proline-rich proteins, in the formation of the cornified cell envelope which performs the main barrier function of the skin.

Recessive loss-of-function mutations in TGM1 have been shown to cause lamellar ichthyosis, a disease characterized by excessive scaling and shedding of the outer epidermis.<sup>[5]</sup>

Histological analysis shows the level of blistering in PSS at the junction of the stratum granulosum and the stratum corneum hence differentiating it from epidermolysis bullosa, in which blistering occurs in the basal keratinocytes of the epidermis or in the upper papillary dermis.

#### **IV. Conclusion**

A keen observant eye with aid of family history and histopathological examination can easily diagnose peeling skin syndrome. Counselling and emollients play a major role in this life long entity.

#### **V. Declaration Of Patient Consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

#### **VI. Source Of Financial Support And Sponsorship**

Nil.

#### **VII. Conflict Of Interest**

Nil

#### **Acknowledgement**

Nil.

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