Peeling Skin Syndrome in a 3 Year Old Female Child- A Rare Genodermatosis

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Abstract

We report a case of 3 year old Hindu female child who presented in skin OPD with complaint of spontaneous peeling of skin from all over the body since birth.

Keywords: Skin peeling syndrome, TGM5 gene, Acral skin peeling syndrome.

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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 [1]. Clinically characterised by painless, continual and spontaneous skin peeling since birth associated with itching, redness and formation of blisters.

Generalized Peeling skin syndrome has been subclassified into a noninflammatory type type A which manifest with white scaling, painless and easy removal of the skin and an inflammatory type B 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 is 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 [2].

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, asteotic skin with fine scaling can be observed over bilateral cheeks, forearms(img 3) and bilateral knees (img 4).

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

History of similar illness in father is present. Skin biopsy was sent from a fresh 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.
Img-1: Erosions present at right ankle with hyperpigmented macules at the dorsum of right foot.

Img-2: Multiple hyperpigmented macules formed as a result of post inflammatory hyperpigmentation present over trunk.

Img-3: Asteotic dry skin over bilateral cheeks associated with fine scaling.

Img-4: Dry asteotic skin over bilateral knees.
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 [3].

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 [2].

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 isodipeptide bonds between adjacent polypeptides [4]. 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 [5].

Histological analysis shows the level of blistering in Peeling skin syndrome 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.

CONCLUSION

In conclusion we report a case of a three year old female child with history of spontaneous peeling of skin from all over body since birth associated with dry asteotic skin. Positive family history present in father. General improvement in condition with age.

Histopathological exam of the skin biopsy showed split at the level of stratum granulosum and corneum. Hence clinical and family history as well as microscopic picture is in favour of the diagnosis of peeling skin syndrome type B.

REFERENCES
1. Kurban and Azar 1969; Levy and Goldsmith 1982; Abdel-Hafez. 1983; Silverman. 1986; Mevorah. 1987; Judge. 2004.
2. Mevorah B, Frenk E, Saurat JH, Siegenthaler G. Peeling skin syndrome: a clinical, ultrastructural and biochemical study. Br J Dermatol. 1987; 116:117–125
3. Judge MR, McLean WHI, Munro CS. Disorders of keratinization. In: Burns T, Breathnach S, Cox C, Griffiths C (eds) Rook’s textbook of dermatology.2004: 2.
4. Kurban AK, Azar HA. Familial continual skin peeling. Br J Dermatol.1969; 81:191–195
5. Huber M, Rettler I, Bernasconi K, Frenk E, Lavrijsen SP, Ponec M, Bon A, Lautenschlager S, Schorderet DF, Hohl D. Mutations of keratinocyte transglutaminase in lamellar ichthyosis. Science.1995; 267:525–528.