CASE REPORT

Familial Peeling Skin Syndrome

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ABSTRACT

Peeling skin syndrome (PSS) is a rare form of icthyosis with an autosomal recessive inheritance that exhibits superficial painless, continual, or seasonal exfoliation. The syndrome generally appears at birth or in infancy. A 12 year old female child born of consanguineous marriage presented with generalized periodic peeling of skin since 2 years of age. It was associated with itching, seasonal variations and erythema. The child also had mental retardation, attention deficit hyperactivity syndrome and ocular albinism. Her younger brother was also affected from the similar disease.

Keywords: Deciduous skin, keratolysis exfoliativa congenita, familial peeling skin syndrome.

INTRODUCTION

Peeling skin syndrome is a rare and poorly classified genodermatosis with different clinical picture, and is mainly characterized by spontaneous separation of the stratum corneum[1, 2]. Histologically, the separation is intracorneal or subcorneal in the peeling skin syndrome [1, 2]. This syndrome can be sporadic or familial, and it has no ethnic predominance[3]. The onset of symptoms is usually at birth or shortly thereafter, but it has also been reported that the onset may occur during adulthood too[1, 4]. It is also known as keratolysis exfoliativa congenita[6], deciduous skin[7] and familial continual skin peeling[8]. PSS is divided into two main types, the generalized and the localized forms. Generalized PSS is divided in three types: type A and type B were identified by Traupe and type C by Mevorah et al.[8]. To date, no effective treatment for peeling skin syndrome has been reported. Treatment with topical corticosteroids, UVB, methotrexate, etretinate, and isotretinoin was found to be ineffective[9].

CASE REPORT

A 12 year old female child reported with history of generalized painless peeling of skin since 2 years of age. She was the first child of her family born of consanguineous marriage by normal vaginal delivery. Initially peeling involved the trunk but later, upper and lower limbs were also involved. It was associated with generalized itching. Worsening of exfoliation of skin was noticed in summer and on exposure to humidity. Dry scaling of the trunk and lower limbs was also noticed since the beginning of the disease. Patient also had poor vision, behavioral problems, stunted growth and delayed developmental mile stones since birth. Both her parents were normal. There was no history of such disease previously in the family. Patient has one younger sister and one younger brother. Her younger brother also developed similar cutaneous lesions since the age of 2 years. Peeling involved his trunk and lower limbs. Patient's younger sister is mentally retarded but has no cutaneous abnormalities.

Cutaneous examination of the patient revealed peeling of skin in form of sheets involving the trunk, flexor and extensor surfaces of upper and lower limbs(figure 1and 2) and face leaving behind erythematous patches with a peeling border. No tenderness, bleeding or discharge was present at the sites of lesions. Scalp, flexures, palms and soles were spared. Hair, nails and oral cavity was also normal. Pediatric evaluation showed both height and weight below the 5th percentile of age. Psychiatric evaluation revealed mental retardation and attention deficit hyperactivity syndrome. On ophthalmological examination the fundoscopy showed normal disc, enlarged choroidal vascular markings, attenuated blood vessels and hypopigmentation of the fundus and iris depicting ocular albinism.

Histopathological examination of cutaneous punch biopsy revealed subcorneal splitting, papillomatosis, elongation of rete ridges, hyperkeratosis and focal parakeratosis in the epidermis. Dermis showed mild mononuclear infiltrate.

Complete blood count, complete urine examination, liver and renal function tests were within the normal limits. She was treated with emollients and antihistamines, after which there was symptomatic relief but on follow up there was recurrence of lesions. History, clinical features and histological picture supported the diagnosis of familial peeling skin syndrome.

DISCUSSION

The peeling skin syndrome is an autosomal recessive disorder characterized by lifelong peeling of the stratum corneum. The cause of this disorder is unknown. The defect is reduced adherence of ab-

normally thick stratum corneum to the stratum granulosum. Abnormal deposition of lipid causes disturbance of skin barrier function and cohesion, which leads to the desquamation of stratum corneum[10]. Rare features of peeling skin syndrome include easily removed hairs, koilonychia, distal onycholysis, chapping, and keratoderma[1]. There is usually no systemic abnormality in this syndrome. However, a few case reports state mental retardation, sexual dysfunction, anosmia, short stature, primary amenorrhea, and sexual infantilism[1, 9]. In our case ocular albinism and attention deficit hyperactivity syndrome was present which has not yet been reported in any case of this disease. In addition patient was also mentally retarded. The skin peeling could be either continuous or periodic, and in some cases, seasonal changes have been reported. In most cases of peeling skin syndrome, the separation of the skin is usually generalized. However, in recent years, some patients with localized acral peeling skin syndrome have been reported as well[1, 2, 11]. The palms and soles may be hyperkeratotic but are spared from peeling in most cases with generalized peeling skin syndrome. Our patient had periodic and generalized skin shedding and showed sparing of palms and soles.

Histological examination may be helpful for the diagnosis by showing superficial cleavage in the epidermis. It is usually localized in the interface of stratum corneum and stratum granulosum and more rarely in the stratum lucidum. In our patient the epidermal separation is subcorneal.

The literature distinguishes two main clinical presentations of PSS i.e. the generalized and the localized forms. Generalized PSS is usually divided in three subtypes: type A, B and C. Type A and type B were identified by Traupe and type C by Mevorah et al.[8]. PSS-type A includes asymptomatic and noninflammatory exfoliation, and general health usually remains intact. It may consist of some hyperpigmentation, but no vesiculation or erythema. PSS-type B includes seasonal variation or periodic peeling with inflammatory changes and pruritus. It shows erythematous plaques with rare vesiculation [12]. PSS-type C usually begins in infancy with erythematous patches surrounded by a superficially peeling collarette. Marked blepharitis, conjunctivitis and cheilitis were observed. Atopy and pruritus were also noted[14]. The peeling begins at birth or at 3 to 6 years of age in type A, and at birth in type B[12]. Our case most likely belongs to type B of generalized PSS as the patient showed periodic peeling of skin with inflammatory changes and pruritus but the peeling started at 2 years of age and not at birth. Patient also showed features of mental retardation and ocular albinism.

Treatment of PSS remains symptomatic. Patient education regarding avoidance of trauma in involved regions, prolonged immersion in water and using absorbing powders and aluminium antiperspirants is helpful. Keratolytics and urea creams speed up shedding. Topical and oral steroids, emollients and tar, oral retinoids, methotrexate and UVB phototherapy are among those agents tried without success[14]. In our patient emollients and anti histamines were given with a temporary response.

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