

Case Report

A case of Kindler syndrome with florid scabies

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Abstract Kindler syndrome is a rare autosomal recessive disorder characterized by congenital blistering and photosensitivity combined with progressive poikiloderma and cutaneous atrophy. The genetic defect has been localized to chromosome 20 and the syndrome results due to mutations in the KIND 1 gene. We report the case of a 12-year-old boy with classical features of blistering since infancy, progressive poikiloderma, photosensitivity and characteristic atrophic cigarette paper-like appearance of the skin. In addition he had florid scabies with quite a few burrows on palms and in finger webs and numerous papular and pustular lesions on trunk and genitalia.

Key words

Kindler syndrome, Weary-Kindler syndrome, poikiloderma

Introduction

Kindler syndrome (KS) is a rare hereditary disorder characterized by acral blister formation in infancy and childhood, progressive poikiloderma, cutaneous atrophy and photosensitivity.¹ It is recently reported that KS is the first genodermatosis caused by a defect in the actin-extracellular-matrix linkage and the gene was mapped to chromosome 20p12.3.² A novel family of focal adhesion proteins, the kindlins, is involved in attachment of the actin cytoskeleton to plasma membrane and in integrin-mediated cellular processes.^{2,3} Deficiency of kindlin 1 as a result of loss-of-function mutations in the KIND 1 gene causes KS.⁴ More than 100 cases have been reported worldwide.⁵ Persons of any race can be affected and no sex predilection has been documented.^{5,6,7} The hallmark of KS is congenital blistering and photosensitivity,

progressive poikiloderma and atrophic changes seen as cigarette paper-like wrinkled appearance of the skin commonly on trauma prone sites.⁸ Dental abnormalities,⁹ leukokeratosis of buccal mucosa, urethral, anal and esophageal strictures, finger webbing, nail dystrophy and skeletal and eye abnormalities are other reported features.^{5,6,7,8} Early development of actinic keratoses can occur and squamous cell carcinoma of lower lip and transitional cell carcinoma of the bladder have been reported.⁵ Secondary infections of the bullous lesions and cosmetic disfigurement are the common causes of morbidity. We report the case of a 12-year-old boy with classical features of KS who presented to us with florid scabies.

Case report

A 12-year-old boy, born of a consanguineous marriage, presented to the dermatology department Jinnah hospital, Lahore with the complaint of blister formation over acral areas since birth and severe generalized nocturnal itching for the

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Figure 1 Poikiloderma over face and upper trunk.



Figure 4 Atrophic skin over dorsa of feet with crusting over lower legs.



Figure 2 Pigmentation over back with crusting in axillae, lower back and buttocks.



Figure 5 Gingival swelling and dental caries.



Figure 3 Atrophy and crusting on dorsa of hands and in finger webs.

past two months. The blisters were more over the forearms and legs, developed spontaneously, were sometimes pus filled and ruptured to heal with scarring and pigmentation. The blistering tendency was pronounced in infancy and throughout

childhood. Along with this the patient developed complaint of photosensitivity and gradually increasing hyper-pigmentation over the face. The blisters gradually subsided and there have been no blisters over the past one year. For the last two months the patient complained of severe generalized pruritus, more at night and with history of itching in other members of the family. On systemic inquiry there was no complaint of dysphagia or any skeletal abnormalities. There was no history of similar blistering skin disease in the family.

Physical examination revealed poikilodermatous skin over neck, upper trunk (**Figure 1**) and limbs (**Figure 2**). There was cigarette paper like atrophic scarring over dorsa of hands (**Figure 3**) and feet (**Figure**

4) along with diffuse palmoplantar keratoderma. There was hyperpigmentation over face predominantly affecting the forehead, cheeks, and nasal bridge. Nails were dystrophic with subungual hyperkeratosis and onycholysis. Erythematous papular and crusted lesions were present over limbs, trunk and hands. Burrows were seen in the interdigital webs. There was gingival swelling (**Figure 5**) and dental carries. Eye examination was normal.

On investigation, complete blood counts, urine examination, liver function tests were normal. On histopathology, changes were suggestive of poikiloderma atrophicans. The patient was treated symptomatically, was given sun protection and his family was counseled about the disease. The patient and his family members were treated for scabies and the symptoms were relieved completely.

Discussion

First described in 1954 by Theresa Kindler, KS is a combination of features of inherited blistering skin disorders and congenital poikilodermas.¹ In 2003, Siegel et al mapped the disease locus to band 20p12.3.⁴ The lack of specific features in early childhood often lead to difficulty or delay in diagnosis. In the newborn period KS may be difficult to differentiate from variants of Epidermolysis Bullosa. Progressive improvement of photosensitivity and blistering followed by appearance of poikiloderma and cutaneous atrophy with age, help confirm the diagnosis.^{5,6,7} Rothmund-Thomson syndrome has poikiloderma and photosensitivity similar to KS but it is also accompanied by short stature, sparse hair, hypogonadism and cataracts which are not features of KS.¹⁰ Several photosensitivity disorders with

defective DNA repair share cutaneous findings with Kindler syndrome. These include xeroderma pigmentosum, Bloom's syndrome, Cockayne's syndrome and dyskeratosis congenita. However, the classical features of acral blistering are absent. Hereditary sclerosing poikiloderma described by Weary in 1969 is an autosomal dominant disease characterized by progressive poikiloderma in flexural areas, sclerotic bands, poor dentition and occasionally calcinosis. Absence of bullae and photosensitivity clearly distinguish it from KS.¹¹ However, in 1971, Weary *et al.*¹² described a large kindred of patients with widespread eczematous dermatitis, acral bullae in infancy, keratoses, and gradual appearance of diffuse poikiloderma. Several authors have noted the similarity of these patients to the original case described by Theresa Kindler, and some believe that Weary and Kindler are variants of the same disorder.¹³

To facilitate clinicians in the diagnosis of KS a set of clinical diagnostic criteria were proposed by Fischer *et al.* in 2005.¹⁴ (**Table 1**). It was considered that presence of 4 major criteria makes the diagnosis of KS certain. The presence of 3 major and 2 minor criteria makes the diagnosis probable and the presence of 2 major and 2 minor criteria or associated features renders the diagnosis likely. In 2007 Al Aboud *et al.*¹⁵ added to the major criteria facial and neck erythema, telangiectasia and history of an affected child in the family. The proposed criteria provide clinical grounds for the diagnosis and may help clinicians to decide upon the patient's referral for genetic consultation.

Table 1 Diagnostic Criteria for Kindler syndrome

Major criteria

1. Acral blistering in infancy and childhood
2. Progressive poikiloderma
3. Skin atrophy
4. Abnormal photosensitivity
5. Gingival fragility and /or swelling

Minor criteria

1. Syndactyly
2. Mucosal involvement(urethral, anal, esophageal, laryngeal stenosis)

Associated findings

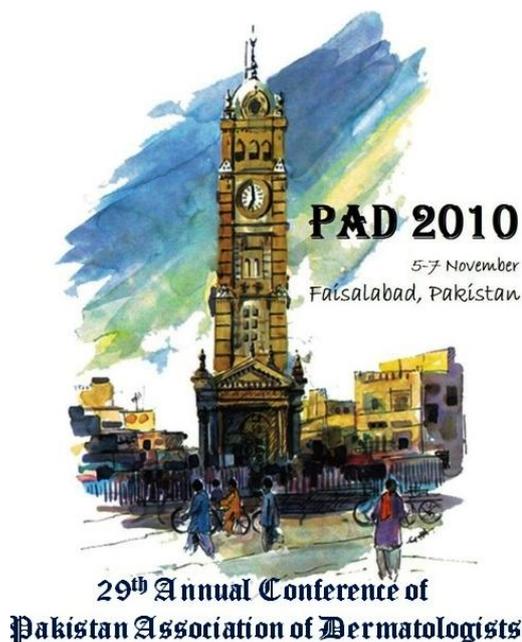
1. Nail dystrophy
2. Ectropion of lower lid
3. Palmoplantar keratoderma
4. Pseudoainhum
5. Leucokeratosis of the lips
6. Squamous cell carcinoma
7. Anhidrosis/hypohidrosis
8. Skeletal abnormalities
9. Poor dentition/dental caries
10. Periodontitis

KS although a rare disorder has been reported before from Pakistan and in Pakistanis settled abroad.¹⁵ The first 3 cases were reported in 1992 by Ahmed *et al.*¹⁶ from Lahore. More recently another case has been reported.¹⁷ Our patient was a 12-year-old boy with classical features of acral blistering since infancy, progressive poikiloderma, skin atrophy photosensitivity and gingival swelling. In the associated features he had nail dystrophy, palmoplantar keratoderma and dental caries. He thus fulfilled all the major criteria hence making the diagnosis certain. In addition he also had florid crusted scabies. It is proposed that the fragility of his skin contributed to the impetiginisation and eczematization with which he presented. We report this case for its association with florid scabies that has not been previously reported.

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