

Kindler syndrome with typical clinical manifestations: A case report from Syria

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ABSTRACT

Kindler's syndrome is a highly rare hereditary disorder, a form of epidermolysis bullosa, caused by a mutation in the KIND1 gene, which is characterized clinically by photosensitivity, poikiloderma, trauma-induced blistering that begins at birth, skin atrophy, mucosal involvement, and a risk of malignancy. Herein, we present a case of Kindler's syndrome and highlight the classical manifestations and associated features of this rare syndrome and diagnosis methods, differential diagnosis, and the necessity of a multidisciplinary team to control the symptoms and improve the patient's quality of life.

Key words: Kindler's syndrome, Photosensitivity, Blistering, Fragility, Poikiloderma, Onycholysis

INTRODUCTION

Kindler's syndrome (KS) is a rare autosomal recessive condition. This genodermatosis, which was first described by Theresa Kindler in the 1950s and classified as a rare form of epidermolysis bullosa, is the outcome of a mutation in the KIND1 or FERMT1 genes. In turn, this mutation makes the skin susceptible to increased photosensitivity, blistering, and fragility [1]. Throughout the world, there are only about 250 cases reported [2,3]. This syndrome is considered to be a combination of manifestations of congenital poikiloderma and inherited blistering skin disorders [3,4].

KS may affect persons of any race and may affect both men and women with no sex predilection [5].

The disease's severity may range from mild symptoms such as blisters and skin ulcers, to more severe symptoms with mucosal involvement, severe esophageal stenosis, anemia, and rarely, colitis [6].

Herein, we report the case of a 39-year-old woman with KS and no familial history.

CASE REPORT

A 39-year-old unmarried woman presented to the dermatology department with complaints of photosensitivity and poikiloderma on sun-exposed areas (face, neck, and V area of the upper chest) (Figs. 1a and 1b). Throughout her life, she suffered from recurrent blistering after minor trauma beginning after birth. The changes were more prominent on the extremities and tended to disappear after the age of 13 years. Subsequently, discoloration and atrophy of the skin developed. Also, she had palmoplantar hyperkeratosis (Fig. 1c), in addition to swallowing difficulties with solid foods and oral medications because of esophageal strictures. Cutaneous examination revealed atrophic, dry skin with cigarette paper-like wrinkling, diffuse poikiloderma (atrophy, telangiectasia, and reticular pigmentation) mainly on the sun-exposed areas (Figs. 1a – 1d). There was also eye redness, ectropion, actinic cheilitis with reduced ability to open the mouth, gingivitis, periodontitis (Fig. 1e) and nail changes, including subungual hyperkeratosis, onycholysis, and transverse and longitudinal ridges (Fig. 1c).

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Figure 1: (a and b) Hypo- and hyperpigmented macules with telangiectasia and photosensitivity on the patient's face, neck, and V area of the upper chest. (c) Atrophic skin with cigarette paper-like wrinkling of the skin. Nail changes, including subungual hyperkeratosis, and onycholysis. (d) Absence of dermatoglyphics. (e) Erosions in the buccal mucosa, angular cheilitis with poor oral hygiene and periodontitis.

Two skin punch biopsies were taken from the forearm and lower leg. Histopathology of these biopsies showed epidermal atrophy with hyperkeratosis, focal liquefaction degeneration of the basal layer with pigmentary incontinence (Fig. 2a). There was dermal edema with dilated blood vessels and a patchy perivascular mononuclear inflammatory cell infiltrate (Fig. 2b).

Based on the patient's history, clinical features, and histopathological findings, Kindler syndrome was diagnosed. For confirmation, genetic testing was advised, but because of the financial limitations, the patient could not afford it.

DISCUSSION

Kindler syndrome is a subtype of epidermolysis bullosa that results from a mutation in the *FERMT1* gene that encodes kindlin-1 protein [7]. Intermittent cases are not uncommon [4].

KS has been mainly described in Arab origin, as from Pakistan, Iran, Turkey, and India. It has also been reported in European individuals, as well as Caucasian, Albanian, Italian, and Serbian [3].

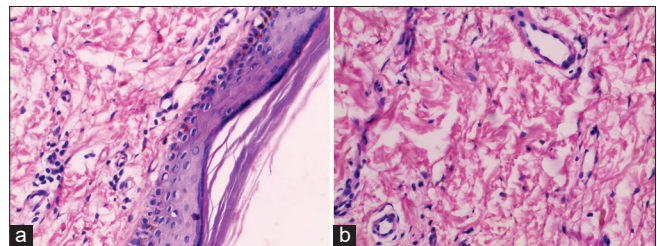


Figure 2: a) Histopathology of the biopsies showing epidermal atrophy, focal vacuolar degeneration of the basal layer with subepidermal cleft (H&E, 100x). b) Dilated blood vessels in the upper dermis (H&E, 200x).

Clinically, KS is characterized by minor trauma-induced blisters at birth or during the first days of child life, and these blisters regress with age. Other features, including photosensitivity, skin fragility, poikiloderma, diffuse skin atrophy, particularly in sun-exposed areas, tend to happen in infancy or early childhood. While photosensitivity becomes better with age, atrophy and poikiloderma worsen over time [3].

KS also has mucosal involvement, which usually begins during adolescence. The most commonly affected mucosa is the oral mucosa, leading to hemorrhagic gingivitis, premature tooth loss,

periodontal disease [2], xerostomia, oral ulcerations, and microstomia [5].

After the age of 10, ocular, esophageal, anal, and urogenital mucosa involvement is more frequent [2].

The ocular features of this disorder include kerato-conjunctivitis, cicatricial ectropion, blepharitis, recurrent corneal erosions, corneal ectasia, pigment deposits over the lens capsule, symblepharon, segmental chorioretinal atrophy, and conjunctival scarring [7].

The patients with KS have an increased risk of developing squamous cell carcinoma of the skin after the age of 45 [2].

The histopathology of Kindler syndrome shows poikilodermatous changes [4].

Numerous diseases that may result in cutaneous atrophy, blistering, and/or poikiloderma should be differentiated from KS, [4] including Weary syndrome, Bloom syndrome, Rothmund–Thompson syndrome, Cockayne syndrome [8], X-linked recessive epidermolysis bullosa simplex, dyskeratosis congenita, Xeroderma pigmentosum, and dermatopathia pigmentosa reticularis [9].

It has also been proposed that KS and Weary’s HAP are variants of one condition themselves [4]. However, the pattern of inheritance of Weary is autosomal dominant, while it is autosomal recessive in KS [9,10]. Also, the occurrence of blistering, photosensitivity, and the presence of eczema is not the same in these two syndromes [10]. In Kindler syndrome, photosensitivity is evident while, in Weary syndrome, there is no photosensitivity. Also, blisters begin arising within the first six months of life in Weary syndrome, unlike KS, where blisters begin at birth, in addition to widespread dermatitis in Weary syndrome, which is absent in KS [9,10].

It might be difficult to distinguish KS from variants of epidermolysis bullosa in newborns. However, in KS, there is the progressive improvement of photosensitivity, blistering, poikiloderma, and cutaneous atrophy over time, unlike epidermolysis bullosa [4].

The associated manifestations in Rothmund–Thomson syndrome, an autosomal recessive disease, such as hypogonadism, sparse fine scalp hair, alopecia,

microdontia, and rarely, mental retardation, are different from KS [9,10].

There is no true poikiloderma in Bloom syndrome, contrary to KS. In Cockayne’s syndrome, associated features, including dwarfism, progressive pigmentary retinopathy, cachexia, deafness, and birdlike faces, help to distinguish this syndrome from KS. In dyskeratosis congenita, unlike KS, the pigmentary alterations are not truly poikilodermatous, and bullae are not an essential feature of this uncommon genodermatosis [4].

In 2004, Angelova-Fischer et al. presented clinical criteria to facilitate the accurate diagnosis of this syndrome (Table 1).

Our patient had met all major criteria, one of the minor criteria, and six of the associated findings.

According to the suggested criteria, diagnosis is confirmed by the presence of four major criteria, diagnosis is probable by the presence of three major and two minor criteria, and diagnosis is likely when two major and two minor/additional features are present [10].

The diagnosis of KS may also be established by immunofluorescence mapping and/or detecting FERMT1 gene mutations [3].

Actually, there is no confirmed treatment for KS, and the management is mainly symptomatic and preventive [4,8].

The patient should avoid sun and heat exposure, apply sunscreens and moisturizers, observe skin malignancies,

Table 1: Diagnostic criteria for Kindler syndrome [8]

Major Criteria
Acral blistering in infancy and childhood
Progressive poikiloderma
Skin atrophy
Abnormal photosensitivity
Gingival fragility and/or swelling
Minor Criteria
Syndactyly
Mucosal involvement (urethral, anal, esophageal, laryngeal stenosis)
Associated Findings
Nail dystrophy
Ectropion of the lower lid
Palmoplantar keratoderma
Pseudoainhum
Leukokeratosis of the lips
Squamous cell carcinoma
Anhidrosis/hypohidrosis
Skeletal abnormalities
Poor dentition/dental caries
Periodontitis

and receive nutritional and psychosocial support [3,8]. The patient should also avoid skin trauma, take care of wounds, and treat infected bullous lesions and ulcerations with topical and systemic antibiotics to minimize the morbidity, which results from secondary infections of blisters, mucosal involvement and leads to anal, urethral, and esophageal stenosis, ocular complications, and aggressive periodontitis [3,4].

The life expectancy of patients with KS is usually normal, but they have a significant risk of developing malignancies, including squamous cell carcinoma on the lip, hard palate, and acral skin, in addition to transitional cell carcinoma of the bladder, which runs a severe course [3].

Patients with Kindler syndrome should be treated with a multidisciplinary team of dermatologists, ophthalmologists, urologists, dentists, gastroenterologists, pediatricians, dieticians, nurse specialists, psychologists, and geneticists [8].

CONCLUSION

Kindler syndrome is an extremely rare disorder. We presented a case with specific features of KS, its associated manifestations, histopathological findings, differential diagnosis and management and aimed to increase understanding of the classical clinical manifestations of KS to facilitate the diagnosis and minimize the need for invasive investigations, as well as ensure that treatment is made by a multi-specialization team.

Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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