Blistering and Mottled Pigmentation in a Young Male

Anupama Bains, Deepak Vedant[1], Abhishek Bhardwaj, Poonam Elhence[1]

Departments of Dermatology and [1]Pathology, AIIMS, Jodhpur, Rajasthan, India

A 21-year-old male born of a non-consanguineous marriage presented with history of blister formation over the extremities and trunk since childhood. The lesions used to rupture in 1–2 days followed by crusting and healing with hyperpigmentation in 7–10 days. Lesions were recurrent and aggravated by trauma. The severity of blistering gradually improved with age. He also had a history of photosensitivity. At about 4 years of age he developed thinning of skin over the dorsa of hands. Diffuse mottled pigmentation was present on the neck and face since 10 years of age. There was no history of oral ulcers, difficulty in urination, defecation, dysphagia, and dysphonia. There was history of similar lesions in his brother. On cutaneous examination, there were multiple erosions over the trunk and extremities with crusting and few vesicles [Figure 1a]. Nikolsky's sign was negative. There was presence of poikiloderma over the neck and mandibular area of the face [Figure 1b]. Over the dorsa of hands and feet, atrophic, cigarette-paper-thin skin was present [Figure 2]. Mucosa, hair, and nails were normal. Skin biopsy from bullous lesion showed subepidermal vesicle filled with lymphocytes, few eosinophils, and proteinaceous fluid [Figure 3a]. Skin biopsy from the poikilodermatous skin showed thinned-out epidermis with loss of rete ridges. Superficial dermis showed dilated capillaries, melanin pigment incontinence, along with mild chronic inflammatory infiltrate [Figure 3b].

Question

What is your diagnosis?

Answer

Diagnosis: Kindler syndrome

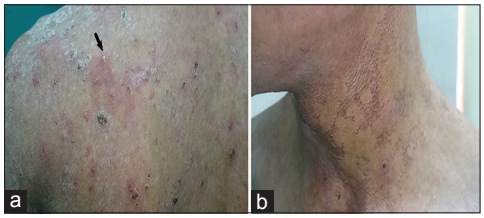

Figure 1: (a) Multiple crusted lesions with black arrow showing vesicle over the back. (b) Poikilodermatous skin over the neck

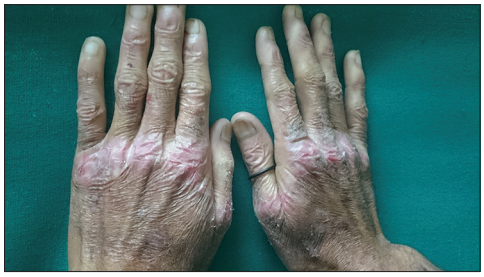

Figure 2: Atrophic cigarette-paper-thin skin over the dorsa of hands

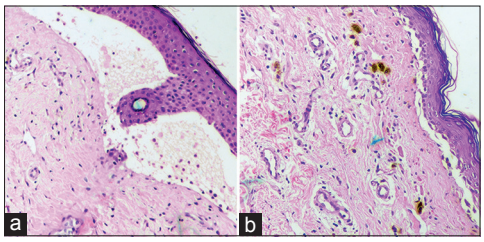

Figure 3: (a) Skin biopsy from bullous lesion showed subepidermal vesicle-filled with lymphocytes, few eosinophils, and proteinaceous fluid (H and E, ×10). (b) Skin biopsy from poikilodermatous skin showed thinned-out epidermis with loss of rete ridges. Superficial dermis showed dilated capillaries and melanin pigment incontinence. (H and E, ×10)

Address for correspondence:
Dr. Anupama Bains, Department of Dermatology, AIIMS, Jodhpur - 342 005, Rajasthan, India.
E-mail: whiteangel2387@gmail.com

Access this article online

Website: www.idoj.in

DOI: 10.4103/idoj.IDOJ_245_17

Quick Response Code:

How to cite this article: Bains A, Vedant D, Bhardwaj A, Elhence P. Blistering and mottled pigmentation in a young male. Indian Dermatol Online J 2018;9:467-8.

Received: September, 2017. **Accepted:** November, 2017.

Discussion

Kindler syndrome is a subtype of epidermolysis bullosa and is characterized by acral blistering, photosensitivity, atrophy, and poikiloderma.[1] It is an autosomal recessive disorder where the defect lies in the *FERMT1* gene on chromosome 20p12.3.[2] The gene encodes for Fermitin (formerly Kindlin-1).[1,3] Kindlin-1 is expressed in basal keratinocytes and has a role in actin-extracellular matrix linkage.[4] Blistering usually starts at birth or within a few days of life and is aggravated by trauma. Photosensitivity is a common finding.[1] Both blistering and photosensitivity tend to improve with age whereas atrophy and poikiloderma are more pronounced in adults, especially in cases with prolonged sun exposure.[2] Cigarette paper-like wrinkled skin is characteristically present in the dorsa of hands and feet.[5] Physical and mental development is normal.[6] Other findings which can be seen are palmoplantar keratoderma; dental caries; periodontitis; hypohidrosis; conjunctivitis; corneal opacity; skeletal abnormalities such as dome-shaped skull and bifid ribs; and mandibular abnormalities. Mucosal findings include leukokeratosis, phimosis, esophageal, urethral, and anal stenoses.[1,6] There is an increased risk of squamous cell carcinoma, especially in elderly population.[1] Histopathology is not very characteristic. The level of split can be intraepidermal, junctional, and at sublamina densa. Poikilodermatous skin shows epidermal atrophy, basal cell vacuolation, pigment incontinence, and mild-to-moderate dermal inflammation.[6] In our patient, the diagnosis of Kindler syndrome was made based on the criteria proposed by Angelo-Fischer *et al*.[7] Our patient fulfilled four major criteria [Table 1]. Kindler syndrome should be differentiated from Weary syndrome and hereditary sclerosing poikiloderma. In Weary syndrome, blistering occurs within 6 months of age, and there are atopic eczema-like features and keratotic papules on the hands, feet, elbows, and knees. It is an autosomal dominant disorder with no photosensitivity, and skin atrophy is not prominent. In hereditary sclerosing poikiloderma, poikilodermatous changes are more common in flexures. There is presence of sclerotic bands, palmar and planter sclerosis, clubbing, and calcinosis cutis. There is no blistering and photosensitivity. Other differentials include Rothmund–Thomson syndrome, epidermolysis bullosa simplex with mottled pigmentation, dystrophic epidermolysis bullosa, xeroderma pigmentosa, dyskeratosis congenita, and dermatopathia pigmentosa reticularis.[3,4] Treatment is usually focussed on genetic counselling, sun protection, and measures to prevent trauma.[1]

Table 1: Criteria for diagnosis of 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 the 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/periodontitis

Criteria for diagnosing a case of Kindler syndrome: 4 major criteria - diagnosis is certain, 3 major and 2 minor criteria - probable diagnosis, 2 major criteria and 2 minor criteria or associated findings - diagnosis is likely

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that name and initial will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Maheshwari A, Dhaked DR, Mathur DK, Bhargava P. Kindler syndrome with palmoplantar hyperhidrosis and blonde hair. Indian Dermatol Online J 2015;6:330-2.
2. Krishna CV, Parmar NV, Has C. Kindler syndrome with severe mucosal involvement in childhood. Clin Exp Dermatol 2014;39:340-3.
3. Youssefian L, Vahidnezhad H, Barzegar M, Li Q, Sotoudeh S, Yazdanfar A, *et al*. The Kindler syndrome: A spectrum of FERMT1 mutations in Iranian families. J Invest Dermatol 2015;135:1447-50.
4. Yazdanfar A, Hashemi B. Kindler syndrome: Report of three cases in a family and a brief review. Int J Dermatol 2009;48:145-9.
5. Gupta V, Dogra D, Gupta N, Parveen S. Kindler's syndrome with long thick cuticles and mottled hyperpigmentation. Indian J Dermatol Venereol Leprol 2011;77:66-8.
6. Sharma RC, Mahajan V, Sharma NL, Sharma AK. Kindler syndrome. Int J Dermatol 2003;42:727-32.
7. Fischer IA, Kazandjieva J, Vassileva S, Dourmishev A. Kindler syndrome: A case report and proposal for clinical diagnostic criteria. Acta Dermatovenerol Alp Pannonica Adriat 2005;14:61-7.