Kindler syndrome: a report of two cases

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Dear Editor,

Kindler syndrome is a rare inherited genetic skin condition [1] that has been classified in the epidermolysis bullosa group [2]. The disease was first described in 1954 by Kindler Theresa [3]. It is characterized by various manifestations of trauma-induced skin fragility that include acral blistering, photosensitivity, progressive poikiloderma, and cutaneous atrophy. These clinical features appear in infancy. Blistering gets quite better with age, whereas poikilodermatous and atrophic changes progressively worsen. In addition, mucosal stenosis can be found in these patients. Oral manifestations with the most common occurrence include aggressive periodontitis, desquamative gingivitis, and angular cheilitis [4]. Periodontal disease has early-onset and rapidly progressive properties, leading to gingival recession and tooth loss [5]. Patients also suffer from cutaneous bullae-induced secondary infections. Recent studies have indicated loss-of-function mutations of the FERMT1 gene. This gene is localized on the short arm of chromosome 20 and encodes the kindling-1 protein of keratinocytes, which has a key role in focal cell adhesion [1]. In this paper, we describe two cases of Kindler syndrome in a single family.

A 21-year-old male patient was referred to our private clinic in Zanjan, Iran. He complained about skin photosensitivity, redness, and atrophy in areas of the hands, foot, and neck. He was born to consanguineous parents. The family also had an undiagnosed daughter. Web formation existed between the second and third fingers in the son (Figure 1). The patients' medical history revealed that trauma-induced blisters had appeared in the neck and acral areas within the first year of life. Dysphagia had started in infancy and persisted throughout the years in the daughter but ceased in the son. Physical examination revealed poikilodermatous skin changes over the son's face, neck, and trunk. The palms showed hyperkeratosis with a dry and atrophic texture; nail dystrophy was also noted. Both patients suffered from laryngeal-esophageal stenosis. Cheilitis and limited oral opening were remarkable. Examination of the oral cavity (Figure 2) showed desquamative gingivitis, severe gingival recessions, and bleeding on probing. Based on the

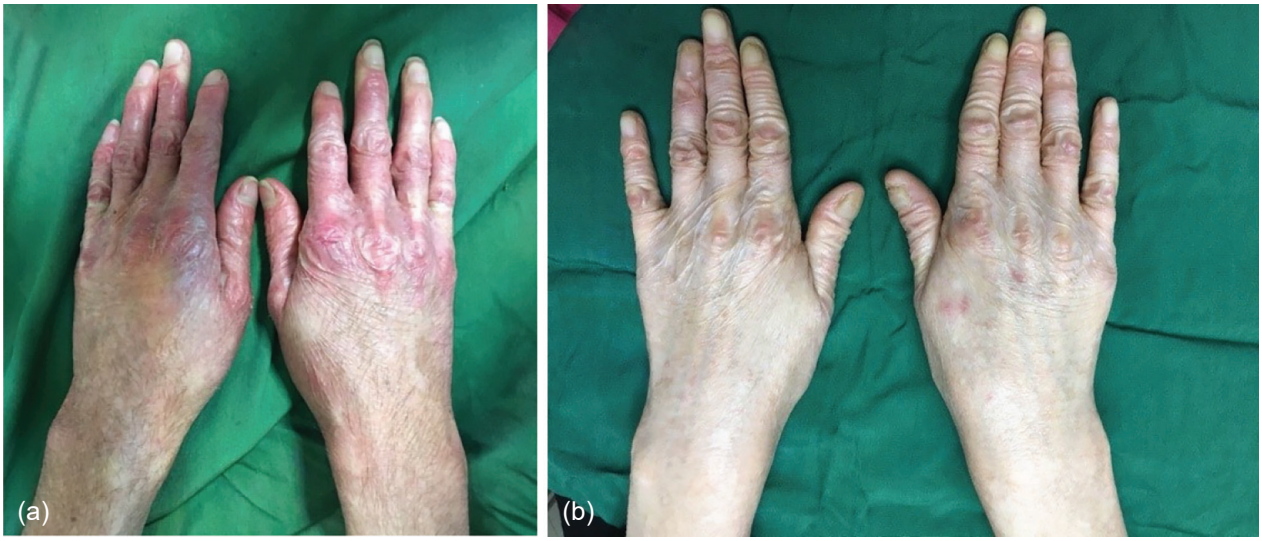

Figure 1. (a) Cigarette paper wrinkling over the dorsa of the hands with dystrophic nails. (b) Web formation between second and third fingers.

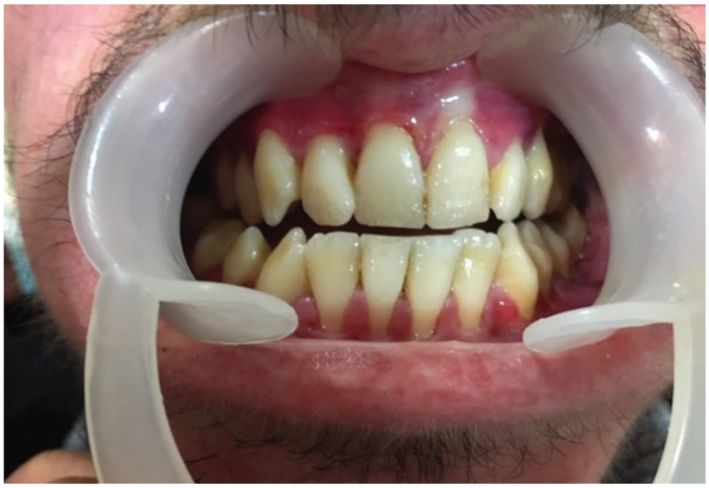
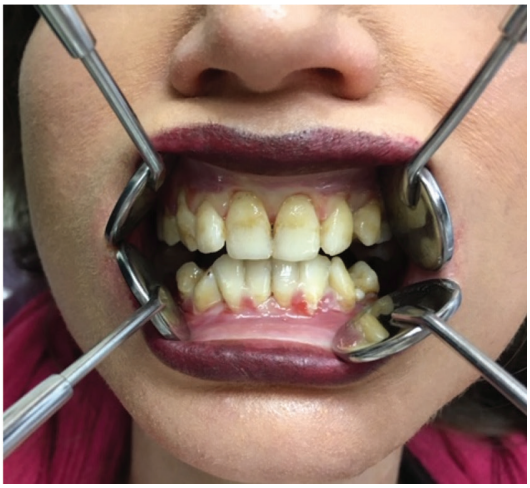

Figure 2. Oral and periodontal status in the patients; severe recession in the gingiva and desquamative gingivitis.

clinical features, our presumptive diagnosis for these siblings was Kindler syndrome. Due to the fact that definitive diagnosis necessitates analysis of gene, mutation analysis of gene with DNA was done after obtaining informed consent. Blood samples were taken and sent to the Thomas Jefferson University. The patients were found to have a homozygous splice site mutation in the non-coding region of the FERMT1 gene. This mutation is inherited in an autosomal recessive manner. In this way, the diagnosis of Kindler syndrome was confirmed. The patients received scaling and root planning together with oral hygiene health instructions. Unfortunately, there currently is no specific therapy for any form of inherited epidermolysis bullosa. Treatment is supportive and the patient should be kept protected from mechanical trauma.

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REFERENCES

1. Gkaitatzi M, Kalloniati E, Has C, et al. Kindler syndrome a rare case report from Greece. Oxf Med Case Reports. 2019;2:103-5.
2. Mendes L, Nogueira L, Vilasboas V, et al. Kindler syndrome-report of two cases. An Bras Dermatol. 2012; 87:779–81.
3. Kindler T. Congenital poikiloderma with traumatic bulla formation and progressive cutaneous atrophy. Br J Dermatol. 1954;66:104–11.
4. Petricca G, Leppilampi M, Jiang G, et al. Localization and potential function of kindlin-1 in periodontal tissues. Eur J Oral Sci. 2009;117:518-27.
5. Yildirim TT, Kaya FA, Taskesen M, et al. Aggressive periodontitis associated with Kindler syndrome in a large Kindler syndrome pedigree. Turk J Pediatr. 2017;59(1):56-61.