



Is adermatoglyphia an additional feature of Kindler Syndrome?*

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DOI: <http://dx.doi.org/10.1590/abd1806-4841.20153501>

Abstract: A typical feature of Kindler Syndrome is skin fragility; this condition is currently classified as a form of epidermolysis bullosa. We describe a rarely reported feature of two cases, one sporadic and one familial; both patients noticed acquired adermatoglyphia. The loss of dermatoglyphics could be an additional feature of this syndrome.

Keywords: Epidermolysis Bullosa; Genetic Diseases, Inborn; Signs and Symptoms

Kindler syndrome (KS; OMIM 173650) is an autosomal recessive genodermatosis characterized by skin blistering, particularly affecting acral sites, with skin fragility and progressive poikiloderma.¹ Mucosal involvement may be present and lead to severe inflammation of the orogenital, ocular and anal mucosa resulting in stenosis and synechiae.¹ Pathogenic mutations in the *FERMT1* gene have been shown to cause molecular KS. The *FERMT1* gene encodes fermitin family homolog (FFH1) protein, also known as kindlin-1, a focal adhesion protein recently implicated in integrin activation in human keratinocytes.² Immunofluorescence microscopy studies show reduced kindlin-1 expression in KS skin and cultured keratinocytes.²

In the current study, we investigated the molecular basis of KS in a female with suspected KS. We also report the unusual finding of adermatoglyphia involving her digits as well as in another confirmed case of KS. The proband is a 26 year-old Brazilian female of African descent who presented with trauma-induced skin fragility, skin atrophy predominantly involving the dorsal surface of hands as well as severe anal, genital and esophageal stenosis. In addition,

she complained that she had been unable to provide adequate fingerprints for official identification purposes in the last few years (Figure 1). On closer inspection, dermatoglyphics were absent on the pulps of her fingers (Figure 1). On the plantar surface of her toes dermatoglyphics were also absent (Figure 1). To investigate whether the patient has KS, a peripheral blood sample was taken from the proband for genomic DNA extraction, polymerase chain reaction amplification of the coding exons and flanking introns of the *FERMT1* gene and bi-directional sequencing as previously described.³ Bi-directional sequencing revealed a homozygous mutation of the *FERMT1* gene, which confirmed the diagnosis of KS. The clinical aspects of this case have already been published.⁴

We then sought to investigate whether other patients with KS have the same clinical finding of adermatoglyphia. Our second case belongs to a large Brazilian pedigree with KS whose clinical details and molecular analysis was published elsewhere.⁵ Briefly, this patient was a 36 year-old male of Italian descent who had skin atrophy, blistering as well as photosensitivity (Figure 2). He also mentioned

Received on 03.03.2014.

Approved by the Advisory Board and accepted for publication on 28.04.2014.

* Work performed at the confocal microscopy outpatient clinic of the Dermatology Department, Santa Casa de Misericórdia de São Paulo – São Paulo (SP), Brazil.

Financial Support: None.

Conflict of Interest: None.

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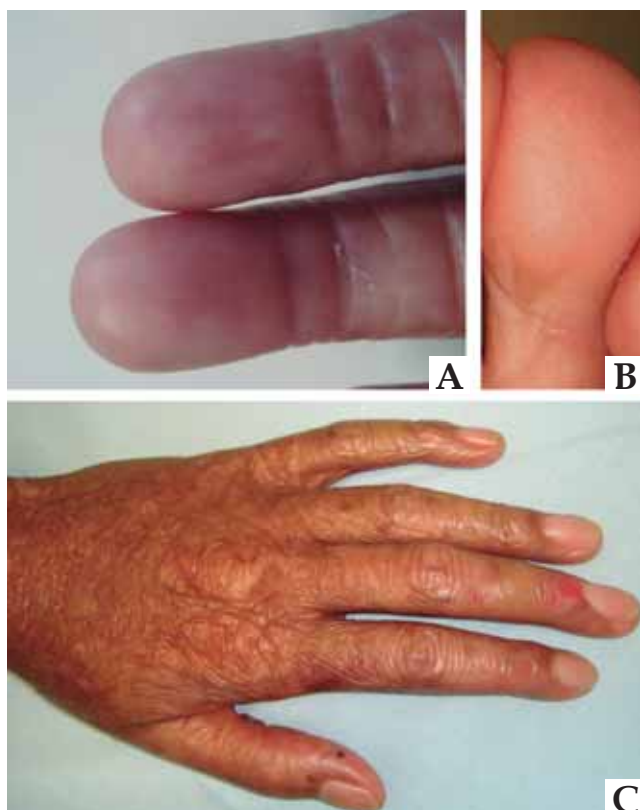


FIGURE 1: First patient with adermatoglyphia of fingers (A) and toes (B). Typical aspect of the dorsum of the hand, note a recent erosion in the middle finger (C)



FIGURE 2: Patient number two with absence of dermatoglyphics (A) and skin atrophy with actinic keratoses (B)

that his fingerprints could no longer be obtained for identification purposes as a result of absent dermatoglyphics (Figure 2).

Adermatoglyphia has previously been described in ectodermal dysplasias and in ADULT syndrome. The first report of acquired adermatoglyphia in KS

was in two German patients, which was described as loss of epidermal ridges.⁶⁻⁸

Probably the typical acral fragility of KS, due to the changes in the dermo-epidermal junction, may lead to secondary absence of dermatoglyphics, which should be regarded as a KS feature. □

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How to cite this article: Almeida HL Jr, Goetze FM, Fong K, Lai-Cheong JE, McGrath J. Is adermatoglyphia an additional feature of Kindler Syndrome? *An Bras Dermatol.* 2015;90(4):592-3.