

Light and transmission electron microscopy of generalized dystrophic epidermolysis bullosa (Pasini's albopapuloid subtype) *

Microscopia óptica e eletrônica de transmissão da epidermólise bolhosa distrófica generalizada (subtipo albo-papulóide de Pasini)

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Abstract: Pasini's albopapuloid epidermolysis bullosa is a very rare subtype of generalized dystrophic dominant epidermolysis bullosa. A 30 year-old white female patient presented since her childhood disseminated small blisters and papules. Light microscopy of a blister showed dermal-epidermal cleavage; moreover, focal areas of dermal-epidermal splitting were also observed. Transmission electron microscopy also identified focal areas of cleavage, which were seen below the lamina densa. It is important to recognize this condition as a variant of epidermolysis bullosa, since the most important cutaneous findings are generalized papules and not blisters and erosions as in other forms of epidermolysis bullosa. **Keywords:** Epidermolysis bullosa dystrophica; Epidermolysis bullosa, junctional; Epidermolysis bullosa simplex; Histology; Microscopy, electron, transmission

Resumo: A epidermólise bolhosa albo-papulóide de Pasini é uma variante rara da forma generalizada de epidermólise bolhosa distrófica dominante. Uma paciente de 30 anos apresenta desde a infância pápulas e bolhas disseminadas. A microscopia óptica de uma bolha demonstrou clivagem dermo-epidérmica; além disso áreas focais de desprendimento dermo-epidérmico foram encontradas. A microscopia eletrônica de transmissão identificou a clivagem abaixo da lâmina densa. É importante que se reconheça essa variante de epidermólise bolhosa, já que o aspecto clínico predominante são pápulas disseminadas e não bolhas como nas outras formas de epidermólise bolhosa.

Palavras-chave: Epidermólise bolhosa distrófica; Epidermólise bolhosa juncional; Epidermólise bolhosa simples; Histologia; Microscopia eletrônica de transmissão

INTRODUCTION

Pasini's albopapuloid epidermolysis bullosa (PAP-EB), described in 1928, is a very rare subtype of generalized dystrophic dominant epidermolysis bullosa (DDEB).¹⁻³ This group of EB has a cleavage under **the lamina densa**, due to dysfunction of the anchoring filaments. Very few cases were reported in the literature.⁴

CASE REPORT

A 30 year-old white female patient was examined in our outpatient clinic. She informed that since her childhood small blisters and papules appear **on** her skin. Some blisters **are** induced by trauma while others **are** spontaneous. Her older sister has similar cutaneous changes.

Skin examination showed disseminated whitish

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papules, which measured a few millimeters. Parallel to these papules a few erosions with hemorrhagic crusts were also observed (Figures 1A and 1B). She presented normal nails, teeth and hairs.

Under light microscopy a dermal-epidermal cleavage was easily identified (Figure 2A). With higher magnification it could be seen that the whitish papules corresponded to superficial dermal elevations, which had higher cellularity and sparse collagen, not arranged in bundles, when compared to the adjacent dermis (Figures 2B and 2C). Scattered in the specimen focal areas of dermal-epidermal splitting were also observed (Figures 3A and 3B). Staining for elastic fibers revealed their absence in these dermal elevations.

Transmission electron microscopy also identified focal areas of cleavage, which were seen below the lamina densa (Figure 4A and B). The dermis beneath the cleavage was formed by loose collagen fibers, which were not forming bundles and by an amorphous material (Figure 4C).

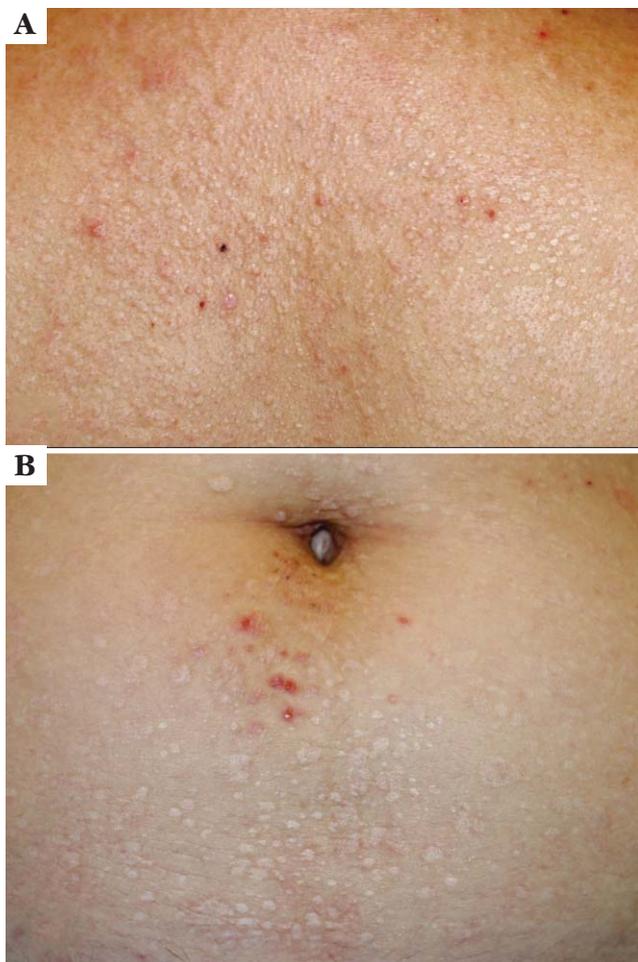


FIGURE 1: Whitish papules and hemorrhagic crusts in the interscapular (a) and periumbilical region (b)

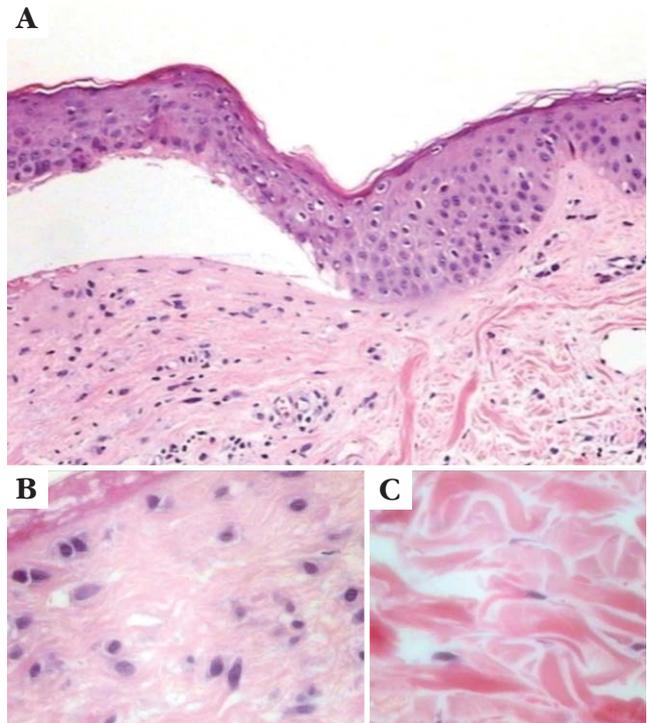


FIGURE 2: Light microscopy: a. dermal-epidermal cleavage (HE x 100). b. detail of the papule with high cellularity and collagen fibers that do not form bundles (HE x400). c. normal adjacent dermis with collagen bundles

DISCUSSION

DDEB is caused by mutations in the COL7A1 gene leading to dysfunction of the anchoring filaments, differently from the recessive group, in which the absence of collagen VII leads to severe clinical features.⁵⁻⁷ Pasini's albopapuloid subtype belongs to the Generalized Dystrophic subgroup, and is characterized by disseminated whitish papules associated with blisters and erosions, as seen in this case.⁸ Nail and mucosal involvement is possible, not found in this patient.

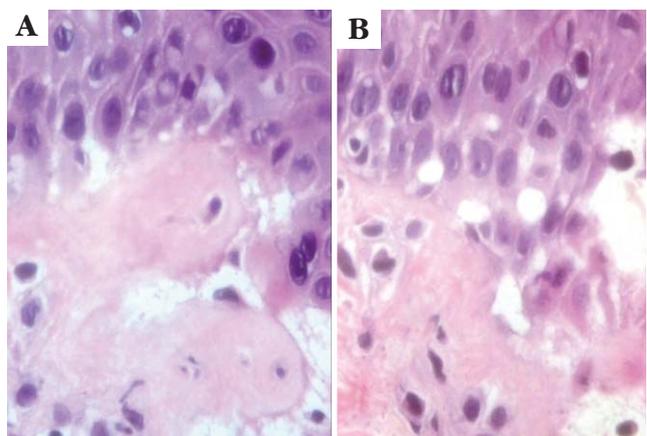


FIGURE 3: Light microscopy: a. and b. focal dermal-epidermal cleavage (HE x 400)

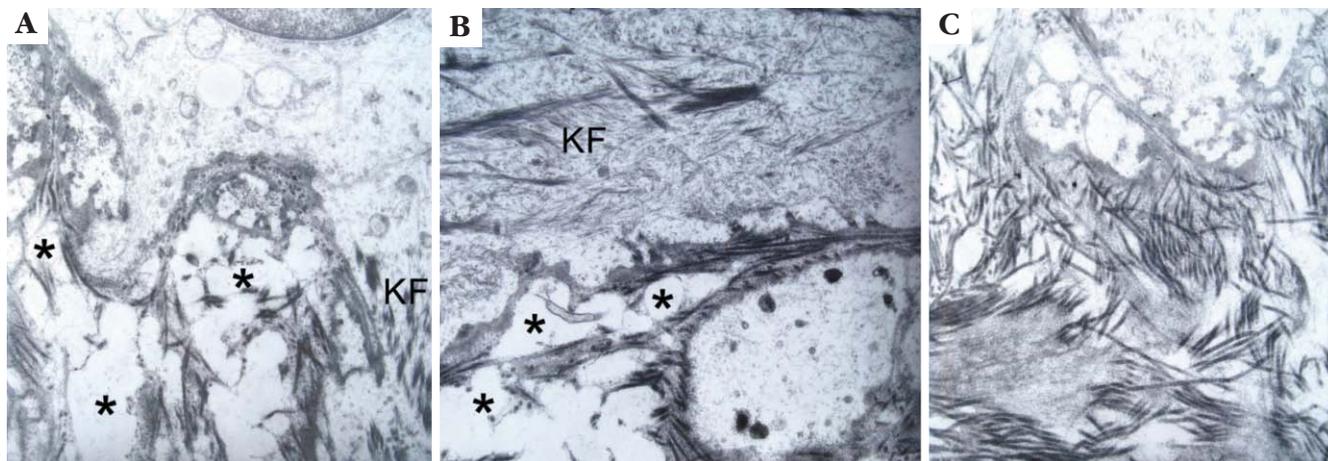


FIGURE 4: Transmission electron microscopy: a. and b. dermal cleavage (asterisks) under the lamina densa (KF – keratin filaments) (x 12.000) c. isolated collagen fibers and deposition of amorphous material in the albopapuloid lesion(x 12.000)

With light microscopy we could demonstrate that the whitish papules were formed by immature collagen with a higher number of fibroblasts, when compared to the adjacent area, according to a previous report.⁹ Moreover, many focal areas of dermal-epidermal cleavage could be seen.

Transmission electron microscopy also showed some areas of focal cleavage, which occurred under the *lamina densa*. Similarly to what was seen with light microscopy, the dermal component of the papule

is formed by isolated collagen fibers. In addition, an amorphous material was also observed, probably a secondary dermal proliferative phenomenon, as described in another case of PAP-EB.⁹

Although this condition has been reclassified as generalized DDEB, it is important to recognize it as a variant of EB, since the most important cutaneous findings are generalized papules and not blisters and erosions as in others forms of EB.⁸ □

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