Qual o Seu Diagnóstico?

A tricoscopia como Pista para o Diagnóstico

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PALAVRAS-CHAVE – Dermatite Esfoliativa; Síndrome de Netherton.

Dermatology Quiz

Trichoscopy as a Clue to the Diagnosis

KEYWORDS – *Dermatitis, Exfoliative; Netherton Syndrome.*

A 43-year-old man presented to our department with generalized erythroderma since early childhood, previously treated with topical and systemic corticosteroids with only partial improvement. The physical examination revealed polycyclic erythematous scaly plaques on the trunk and extremities and lichenification of the body folds (Fig. 1). In addition to skin lesions, we observed sparse hair of both eyebrows and eyelashes, however hair of the scalp was normal. The patient had a history of asthma in childhood. The family history was non-contributory and the patient didn't have any children. Analytically, routine blood tests were normal, however an elevated serum level of IgE 29650 UI/mL (N < 165) was detected.

We performed trichoscopy of the eyebrows that showed nodules along the hair shaft and distal fractures (Fig.s 2a and 2b). The trichoscopic examination of the hair of the scalp did not identify any changes.

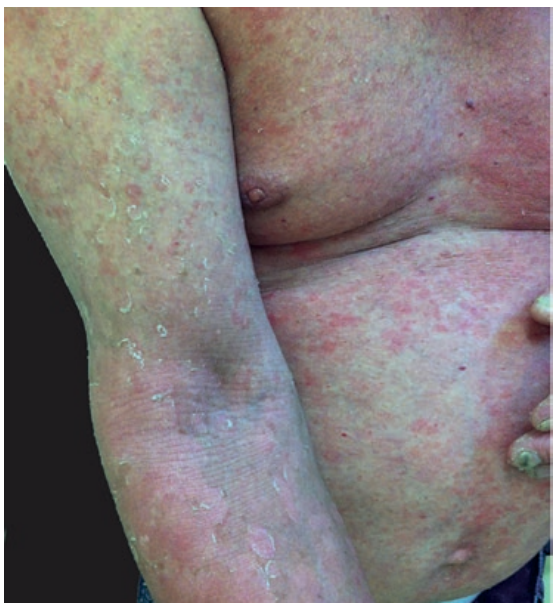

Figure 1 - *Ichtyosis linearis* circumflexa: Polycyclic erythematous scaly plaques on the trunk and upper extremities.

Apresentações: Apresentado sob forma de poster na Reunião da Primavera de SPDV, 2016.
Presentations: Presented as a poster at the Spring Symposium of the Portuguese Society of Dermatology and Venereology, 2016

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Recebido/*Received*
28 Abril/*April* 2017
Aceite/Accepted
27 Junho/*June* 2017

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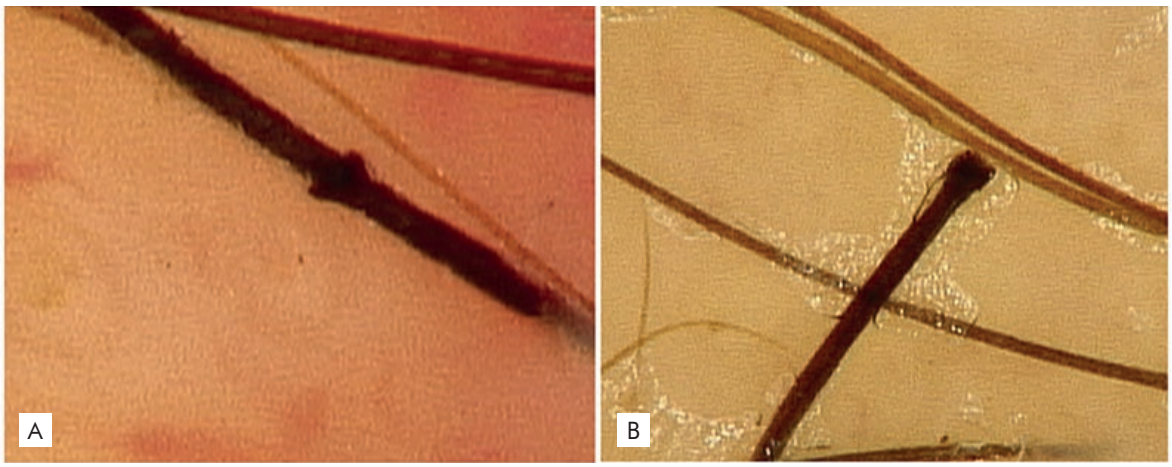

Figure 2 - Trichoscopy: nodules along the hair shaft: "bamboo hair" (**A**) and hair fractures with distal nodules: "matchstick hair" (**B**).

DIAGNÓSTICO:

COMÈL-NETHERTON SYNDROME

Comèl-Netherton syndrome (CNS) is an autosomal recessive genodermatosis, caused by mutation of the SPINK5 gene encoding the lympho-epithelial Kazal-type-related inhibitor (LEKTI). LEKTI is expressed in stratum corneum and its deficiency leads to premature desquamation and severe defect of skin barrier.[1] Clinical features include severe atopy and various allergic manifestations (including elevated IgE and hypereosinophilia), failure to thrive and congenital erythroderma progressing to *ichtyosis linearis circumflexa*.[2,3] Important differential diagnosis of CNS is severe atopic dermatitis, severe eczema, erythrodermic psoriasis and other non-bullous congenital ichthyoses.

Hair is usually sparse and the trichoscopy may show typical abnormalities of the hair shaft, namely *trichorrhexis invaginata*, that is considered pathognomonic.[4] This finding is caused by invagination of the distal part of the hair into the proximal, forming a nodule ("bamboo hair") or a cup-like ending of the hair ("matchstick hair") if fractured. Over the time, cutaneous and hair findings have tendency to improve so adult patients may present only with milder changes and thus pose a significant diagnostic challenge to the clinician.

The reported patient with previously unrecognized CNS presented with normal hair of the scalp and only a careful trichoscopic examination of the eyebrows revealed specific hair changes confirming the diagnosis of CNS. Similarly, Boussofara *et al* described a case of identical twins with CNS presenting with normal scalp hair and *trichorrhexis invaginata* was identified only on the eyebrows. This suggests that the preferred localization of trichoscopic examination is the hair of the eyebrows.[5,6]

As presented, trichoscopy represents a simple tool to visualize TI and thus help establish the diagnosis. The identification of one hair with the typical invagination is sufficient to establish the diagnosis.[6]

Conflitos de interesse: Os autores declaram não possuir conflitos de interesse.
Suporte financeiro: O presente trabalho não foi suportado por nenhum subsídio ou bolsa.
Confidencialidade dos dados: Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.
Protecção de pessoas e animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos responsáveis da Comissão de Investigação Clínica e Ética e de acordo com a Declaração de Helsínquia da Associação Médica Mundial

Conflicts of interest: *The authors have no conflicts of interest to declare.*
Financing Support: *This work has not received any contribution, grant or scholarship.*
Confidentiality of data: *The authors declare that they have followed the protocols of their work center on the publication of data from patients.*
Protection of human and animal subjects: *The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).*

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