

CASE FOR DIAGNOSIS

Linear Hyperkeratotic Papules in a Full Term Newborn



Pápulas hiperqueratóticas con disposición lineal en un recién nacido a término

We present a male newborn, was the second of non-consanguineous healthy parents, born from a full-term spontaneous delivery. At birth, he presented cutaneous lesions in a Blaschko-linear distribution. The affection was extended in the right half of the body, spreading into the trunk, extremities and genitalia characterized by whitish keratotic tiny papules, and in soles and palms were forming hyperkeratotic plaques. The mucose were normal (shown in Figs. 1 and 2).

A complete physical examination revealed no other abnormalities. Skin biopsy showed acanthosis, focal hyperparakeratosis filling follicular infundibula and acrosyringia, and thin columns of parakeratosis emerging from the intraepidermal segments of the anexa (shown in Fig. 3). The lesions spontaneously improved in the first month, remaining just the sole, palm and arm. A transfontanelar ultrasound was performed, without abnormalities. The patient continued with normal development and is followed by the dermatology and paediatrician department.



Figure 1 Linear micropapules following the Blaschko lines.



Figure 2 Hyperkeratotic plaques in right sole.

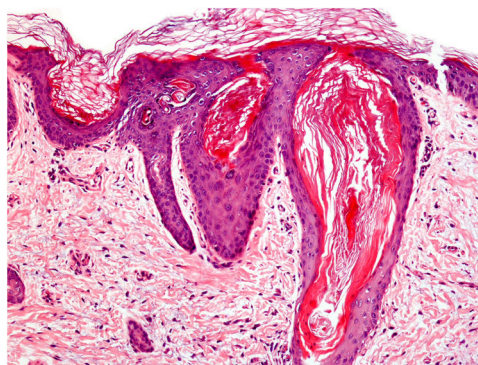


Figure 3 Acanthosis, focal hyperparakeratosis and acrosyringia (H and E, ×20).

What is your diagnosis?

Diagnosis

Porokeratotic adnexal ostial nevus (PAON).

Comments

The diagnosis of this newborn was a Porokeratotic adnexal ostial nevus (PAON). Initially the physical examination with the characteristic distribution of the lesions following Blaschko lines gave us the diagnostic suspicion of this disease. However until the results of the biopsy we did not have the definitive diagnosis.

PAON is a rare benign hamartomatous malformation, usually with a Blaschkoid pattern.¹ It is typically congenital, but it is not always presented in the neonatal period and there are some cases of late-onset PAON. Clinically, PAON present two different types of lesions: palmoplantar papules that present keratotic plugs with central pits, and keratotic papules with a linear distribution in the rest of the body. It is mostly unilateral but there are some cases described of bilateral affection². The exact pathogenesis is unclear but may be produced by a mutation in GJB2 gene, which encoded the connexin 26, a gap junctional protein that participates in keratinocyte growth and differentiation. This mutation is associated with Keratitis-ichthyosis-deafness syndrome, considering PAON a mosaic form of this syndrome. Its inheritance is autosomal dominant although it could express as a de novo mutation. In some patients coexist problems like seizures, hemiparesis, hyperthyroidism, polyneuropathy, scoliosis, deafness or development delay.²⁻⁴ However, the majority of PAON cases present as solitary lesions.⁵ The differential diagnosis includes linear porokeratosis, inflammatory linear epidermal nevus, incontinentia pigmenti, Darier disease, hypomelanosis of Ito and linear lichen striatus among others.

We have to consider this entity in the Blaschkoid pattern of neonatal lesions and make the histopathology study to achieve the diagnosis.

Conflicts of interest

The authors declare that they have no conflicts of interest.

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