DOI: http://dx.doi.org/10.3315/jdcr.2013.1138

Journal of Dermatological Case Reports

Angora hair nevus. A further case of an unusual epidermal nevus representing a hallmark of angora hair nevus syndrome.

María del Carmen Boente[1], *Raúl Asial*[2], *Norma Beatriz Primc*[1], *Rudolf Happle*[3]

1. Department of Dermatology, Hospital del Niño Jesús, San Miguel de Tucumán, Tucumán, Argentina;
2. Universidad Nacional de Tucumán, San Miguel de Tucumán, Tucumán, Argentina;
3. Department of Dermatology, University Medical Center Freiburg, Freiburg, Germany.

Corresponding author:

María del Carmen Boente

Pasaje Bertrés 224,
Tucumán(4000), Argentina

E-mail: *mboente@arnet.com.ar*

Key words:

epidermal nevus syndromes, depigmented hair, hypertrichosis, neurocutaneous syndromes, ocular anomalies, organoid epidermal nevi

Abstract

Background: The association of Blaschko lines with genetic mosaicism has lead to the concept that this pattern represents the manifestation of genetically abnormal skin tissue contrasting with the genetically normal skin. Various mosaic defects affecting not only the skin but also extracutaneous tissues have lead to the description of different types of epidermal nevus syndromes. We present a further case of an unusual organoid epidermal nevus characterized by depigmented hypertrichosis.

Main observations: We describe a 2-year-old boy with a systematized angora hair nevus being characterized by bands covered with soft white hair arranged along Blaschko's lines, involving the scalp, face, and trunk. A biopsy obtained from a scalp lesion showed mild epidermal acanthosis and increased pigmentation of the basal layer. Trichoscopy the affected scalp hair demonstrated fine light coloured shafts. The boy had slight macrocephaly and body asymmetry, a sacral pit, and koilonychia of the big toes.

Conclusion: The angora hair nevus is a peculiar type of organoid epidermal nevus, representing the cutaneous hallmark of a distinctive syndrome, the angora hair nevus syndrome (Schauder syndrome). In cases of epidermal nevi showing hypertrichosis, this unusual entity should be borne in mind for differential diagnosis. (*J Dermatol Case Rep*. 2013; 7(2): 49-51)

Introduction

Epidermal nevi (EN) represent either genomic or epigenetic mosaicism, and most of them originate from post-zygotic mutations.[1-5] EN are further divided into organoid types characterized by hyper- or hypoplastic changes of adnexal structures such as sebaceous glands, sweat glands and hair follicles, and nonorganoid (keratinocytic) types that are characterized by specific changes of the epidermis.[1] It should be borne in mind, however, that an organoid nevus such as nevus sebaceus may likewise show epidermal hyperplasia, and that the organoid component can even be partly absent, especially in those parts of a systematized sebaceous nevus that involves the trunk or the limbs.[2,3]

The epidermal nevus syndromes (ENSs) represent a group of disorders that can be distinguished by the type of associated epidermal nevus and by the criterion of presence or absence of heritability.[1,2] Each type of ENS can be associated with specific abnormalities of the bones, the eyes, the central nervous system, or other organs.[1,2]

Well-defined syndromes characterized by **organoid epidermal nevi** include Schimmelpenning syndrome, phacomatosis pigmentokeratotica, nevus comedonicus syndrome, **angora hair nevus syndrome**, and Becker nevus syndrome. The molecular basis of these disorders has so far not been identified.

By contrast, the group of syndromes characterized by keratinocytic nevi comprises three phenotypes with a known molecular etiology in the form of CHILD (congenital hemidysplasia with ichthyosiform nevus and limb defects) syndrome, type 2 segmental Cowden disease, and fibroblast growth factor receptor 3 epidermal nevus syndrome (García-Hafner-Happle syndrome), whereas Proteus syndrome is still of unknown origin.[1]

We present another case of angora hair nevus, a type of epidermal nevus characterized by **lanugo-like depigmented hypertrichosis** following Blaschko's lines. It represents

the hallmark of a new epidermal nevus syndrome first described by Schauder *et al.*,[6] for which the name angora hair nevus syndrome has been proposed.[1,2]

Case Report

A 2-year-old boy born at term from an uncomplicated pregnancy showed since birth hyperkeratotic hyperpigmented linear lesions on the face, the trunk and the limbs. During his first year of life, he developed patches of light colored hair on his scalp. His parents were healthy and non-consanguineous.

Physical examination showed on the scalp bands of white and rather soft hair alternating with brown colored hair of normal structure (Fig. 1). After shaving these scalp lesions, we exclusively noted dilated follicular pores without other epidermal changes. These areas showed a pattern of distribution following Blaschko's lines of the scalp (Fig. 2A,B). On his face, neck and left ear several bands of hyperkeratotic, hyperpigmented lesions following Blaschko's lines were observed (Fig. 1).

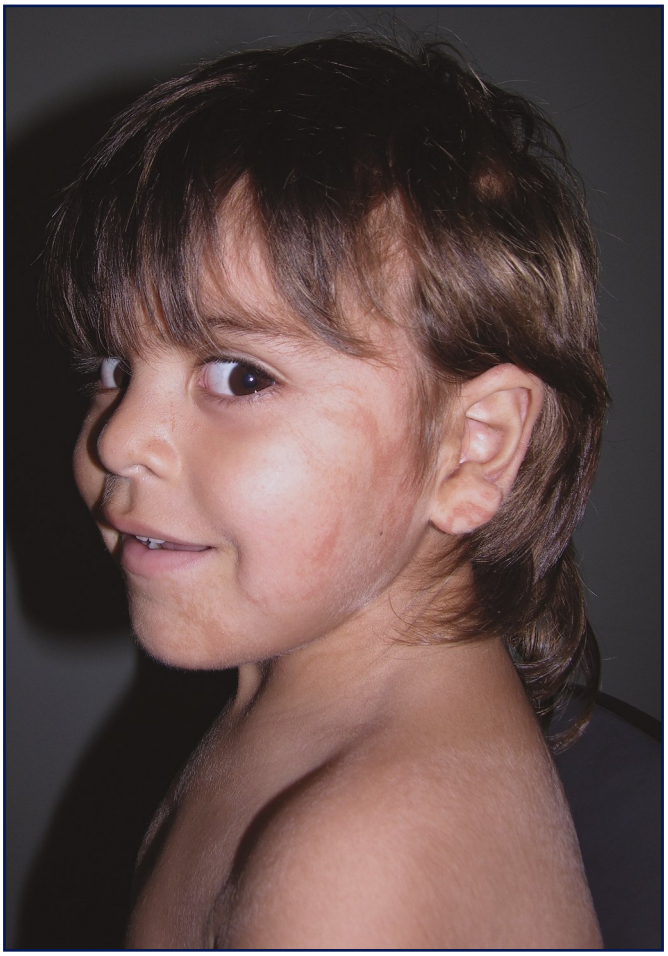

Figure 1

Areas of hypopigmented fine hair on the scalp, and a band of white soft hair involving the left shoulder. Note hyperpigmented and hyperkeratotic linear areas on the left side of the face, and the left ear.

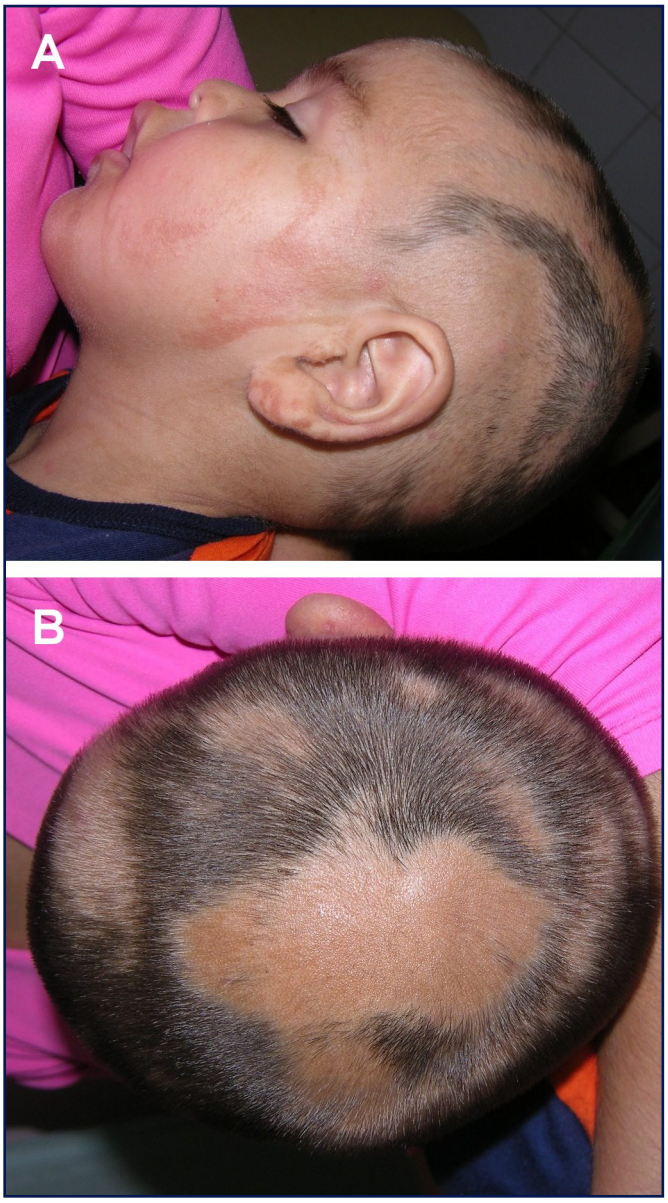

Figure 2

After shaving the scalp: Note Blaschko-linear bands without epidermal changes on the involved scalp skin. A: Hyperpigmented linear bands are observed on the face, ear and neck. B: Note dilated follicular openings in the involved areas. Fine white hair shafts are also observed.

Linear areas of hypertrichosis, covered with white lanugo-like hair, were also noted on his face, trunk, and limbs (Fig. 1).

A biopsy obtained from a scalp lesion showed mild epidermal acanthosis and increased pigmentation of the basal layer.

Dermatoscopic evaluation of the affected scalp hair demonstrates fine light colours shaft without medulla. No other structural abnormalities were seen.

In addition, the boy had slight macrocephaly and body asymmetry, a sacral pit, and koilonychia of the big toes. No ophthalmological, neurological or cardiovascular anomalies were detected.

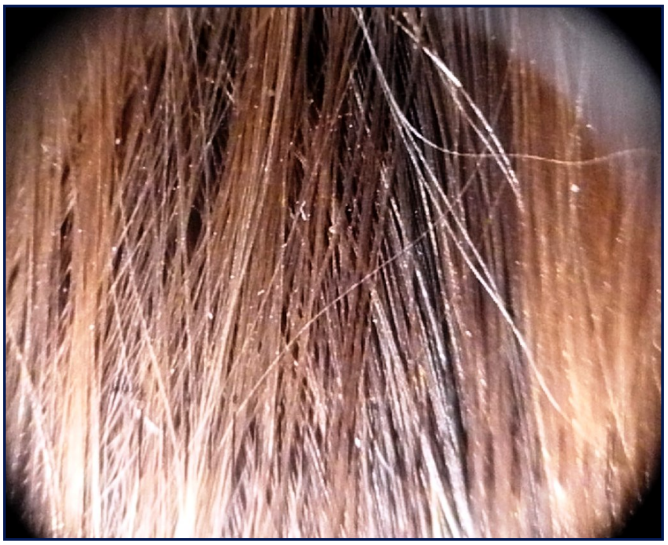

Figure 3

Trichoscopy showed normal hairs shafts, characterized by diversity in color with presence of several gray hairs. Dirty dots – visible in this image – are a normal finding in healthy children.

Table 1. Clinical features of angora hair nevus syndrome.

Cutaneous Findings:
Linear epidermal nevus covered with long, soft white hair. Dilated follicular openings. Linear hyperkeratotic and hyperpigmented areas. Koilonychia. Sacral pit.
Extracutaneous Findings:
Neurologic: Mental retardation, seizures, spastic hemiparesis, dysdiadochokinesis. MRI features: asymmetry of the hemispheres, dilated ventricles, porencephaly
Ophthalmologic: Ectopic pupils, iridocorneal adhesions, iris coloboma, cataract, membranous thickening of the optic nerve heads
Skeletal: Macrocephaly, frontal bossing, body asymmetry
Other features: Macrostomia, malformed ears

Discussion

In 2000 Schauder *et al*.[6] described a new syndrome characterized by an epidermal nevus showing depigmented hair following Blaschko's lines in association with cerebral and ocular anomalies. The hair was fine and lanugo-like, which is why the name **angora hair nevus** has been proposed.[2] This particular organoid nevus distinguishes the syndrome from all other birth defects associated with epidermal nevi.[1]

The angora hair nevus is characterized by rather broad, band-like areas covered with long, smooth, white hair resembling angora hair and growing out from dilated follicular pores.[2] In our case, systematized linear areas of fine, hypopigmented, lanugo-like hair involved the head and the body. Moreover, dilated follicular openings were seen after the scalp hair was shaved. Some of the band-like lesions seen on the face and neck, however, appeared to be hairless and showed acanthosis and slight hyperpigmentation.

The peculiar hypertrichosis associated with angora hair nevus should be distinguished from the woolly hair that may be noted on scalp lesions of systematized non-organoid epidermal nevi.[7] By contrast, sebaceous nevi involving the scalp are characterized by hairlessness.[8]

In the case of Schauder *et al*.,[6] a systematized epidermal nevus showing depigmented hypertrichosis was associated with cerebral and ocular developmental anomalies suggesting a neurocutaneous syndrome reflecting mosaicism. For this phenotype the names **angora hair nevus syndrome** or **Schauder syndrome** have been proposed.[1-2] In our case the systemic involvement was rather mild. With the exception of slight macrocephaly and body asymmetry no other systemic involvement was found (Table 1).

In conclusion, the angora hair nevus appears to represent a distinct type of epidermal nevus. In cases of epidermal nevi showing depigmented hypertrichosis, this unusual entity should be borne in mind and the patient should be examined for the presence of ocular, neurological, skeletal or other extracutaneous anomalies.

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