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Nevus Psiloliparus in a Child with Encephalocraniocutaneous Lipomatosis

Nevo psilolíparo en un niño con lipomatosis encefalocraneocutánea

To the Editor:

Nevus psiloliparus—from the Greek *psilos* ('hairless') and *liparos* ('fatty')—is a term coined by Happle to describe a particular adipose tissue nevus that develops in patients with encephalocraniocutaneous lipomatosis (ECCL).[1] The nevus clinically presents as a slightly raised plaque, with well-defined and sometimes irregular borders, and with slightly yellowish or normal coloring. The unilateral lesion is normally located in the frontal or frontoparietal region, and its most characteristic feature is the paucity or absence of hair follicles.[2] Histologically, nevus psiloliparus has 3 main signs: an abundance of nonencapsulated mature adipose tissue, which can produce compression and thinning of the dermis; a paucity or absence of mature hair follicles; and the presence of normal quantities of orphaned arrector pili muscle bundles (that is, arrector pili muscles independent of or unassociated with hair follicles) arranged in a row parallel to the epidermal surface. The sebaceous glands may be normal or enlarged.

We describe a case of nevus psiloliparus associated with characteristic ECCL traits.

The 2-month old baby boy was born to healthy and nonconsanguineous parents following a normal twin pregnancy with caesarean delivery at term. His identical twin brother was entirely healthy. The patient was brought to the dermatology clinic due to the presence of an area of congenital alopecia on the scalp. Physical examination revealed the right parietooccipital region to have a skin-colored, well-defined, tear-shaped plaque, with a smooth surface and soft consistency, and with reduced capillary density (Figure 1A). In addition, a yellowish papule with well-defined borders and an elastic consistency was observed on the medial third of the right upper eyelid, and a translucent, yellowish plaque with well-defined borders and with telangiectasia was observed at the corneoscleral junction of the right eye (Figure 1B). These signs were suggestive of choristoma of the bulbar conjunctiva. Examination revealed no other lesions, masses, or organomegaly, and no lymphadenopathies in areas accessible to palpation. Biopsy of the scalp lesion showed a normal epidermis, a thinned dermis with a paucity of hair follicles, bundles of smooth muscle arranged parallel to the skin surface, and a disproportionate amount of nonencapsulated mature adipose tissue in the hypodermis (Figure 2). Nuclear magnetic resonance imaging performed when the infant was 3 months old revealed enlargement

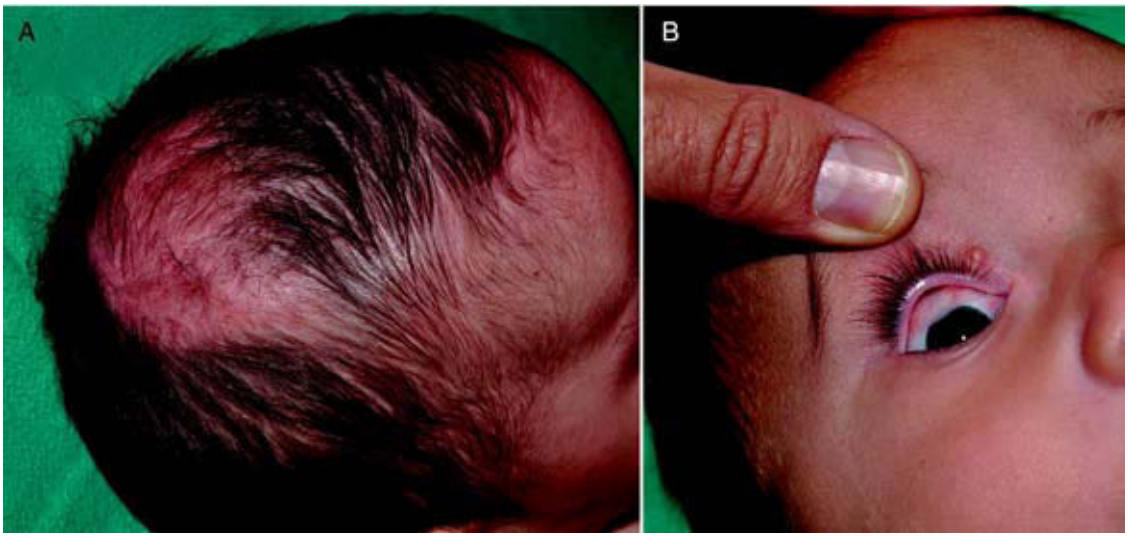

Figure 1 A, Slightly raised and somewhat irregular plaque with well-defined borders, and with a clear reduction in hair follicle density. B, Well-defined, slightly yellowish papule measuring 4 mm in diameter on the medial third of the right upper eyelid, and a translucent, yellowish-pink plaque with superficial telangiectasia extending across the corneoscleral junction.

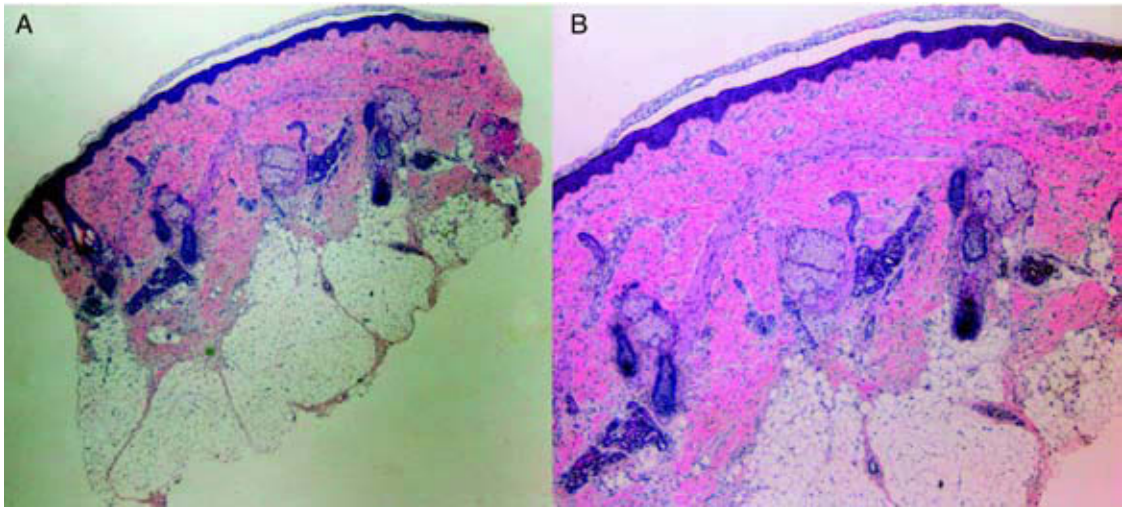

Figure 2 A, Scalp biopsy showing absence of mature hair follicles, horizontal bundles of smooth muscles in the dermis, significant quantities of nonencapsulated mature fatty tissue below the dermis, and a normal epidermis (hematoxylin-eosin, original magnification ×4). B, Higher magnification showing normal quantities of arrector pili muscle bundles parallel to the skin surface (hematoxylin-eosin, original magnification ×10).

of the subarachnoid space in the right frontal, parietal, and temporal regions, and an electroencephalogram performed when the infant was 7 months old showed normal activity. Ophthalmologic examination revealed peripapillary pallor in the fundus of the right eye, and visual-evoked potentials identified an absence of binocular response to a flash stimulus. Ultrasound of the optic nerve revealed no abnormalities of significance. Cardiology and otorhinolaryngology studies were normal. At the time of writing, the psychomotor development of the patient, now aged 11 months, is appropriate for his age.

ECCL is a rare syndrome that shows no association with race, sex, parental age, or a background of consanguinity.[3,4] It is thought to be a lethal autosomal mutation that survives in a mosaic state,[2] and is characterized by the presence of cutaneous, ocular, and neurological lesions, and less frequently, lesions in other organs (Table).

Skin lesions typically affect a single side of the face or scalp, although bilateral involvement has been reported in up to 30% of cases.[5] The lesions may be distributed along a line running from the frontal region to the palpebral region.[2] Nevus psiloliparus is considered to be a hallmark of ECCL and is accepted as a major diagnostic criterion.[5,6] Skin-colored papules may be observed on the forehead or eyelids, corresponding histologically to lipomas,[7] fibrolipomas, angiofibromas,[1] fibroepithelial polyps (skin tags),[8] or choristomas, or may even be similar to nevus psiloliparus.[2] Aplasia cutis congenita has been described as occurring in close association with nevus psiloliparus—possibly a manifestation of non-allelic twin spotting (didymosis aplasticopsilolipara[2]). Although nevus psiloliparus is considered to be a marker for ECCL, it has, on rare occasions, been described as a nonsyndromic skin disorder.[9]

Table 1 Abnormalities encountered in encephalocraniocutaneous lipomatosis[5]

Cutaneous	Ophthalmologic	Neurological	Other
Nevus psiloliparus	Choristomas:	Mental retardation	Macrocephaly
Aplasia cutis congenita	- Dermolipomas	Convulsions	Ossifying fibromas
Lipomas:	- Lipodermoids	CNS abnormalities:	Aortic coarctation
- Zygomatic	Hypertelorism	- Intracranial and intraspinal lipomas	Hypospadias
- Frontotemporal	Retrobulbar tumors	- Arachnoid cysts	Pelvic kidney
Linear papules:	Iris dysplasia	- Hemispheric atrophy	Ear canal stenosis
- Fibromas	Scleral and corneal abnormalities	- Porencephalic cysts	
- Fibrolipomas	Anterior chamber alterations	- Ventriculomegaly	
- Choristomas	Ocular and palpebral colobomas	- Hydrocephalus	
Café-au-lait spots	Aniridia	- Ancephalotrigeminal angiomatosis	
	Microphthalmos		
	Orbital calcification		
	Disrupted eyebrows		

Abbreviation: CNS, central nervous system.

There are numerous ocular and neurological lesions in ECCL, which occur ipsilaterally to the skin lesions. Particularly common, however, are what are called epibulbar choristomas; histologically these choristomas, like the skin lesions, are epibulbar dermoids[10] or fibrolipomas. The severity of associated neurological disorders is important for the prognosis. These disorders, which do not correlate with the extent of the skin lesions[2] or with radiological anomalies,[4] range from no abnormalities at all to convulsions and severe psychomotor dysfunction.

In conclusion, nevus psiloliparus is both a marker for and the first visible sign of ECCL. Meticulous ophthalmologic and neurological examination is crucial for any infant presenting with this particular fatty tissue nevus. Imagining studies should also be performed to rule out associated disorders.

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Specific Palatal Infiltration in B-Cell Chronic Lymphocytic Leukemia

Infiltración específica en el paladar por leucemia linfática crónica B

To the Editor:

Chronic lymphoproliferative syndromes comprise a heterogeneous group of diseases characterized by a monoclonal expansion of mature lymphoid cells. The most common entity in the West is B-cell chronic lymphocytic leukemia, whose tumor cells originate from mature CD5+/CD23+ B lymphocytes present in the bone marrow, peripheral blood, and lymphoid organs. It is typically seen in the elderly, with just 10% to 15% of cases in the under 50s, and usually presents as asymptomatic lymphocytosis though it can sometimes spread to the lymph nodes, spleen, bone marrow, and other tissues.

As in other hematologic neoplasms, nonspecific skin and mucosal lesions can develop in B-cell chronic lymphocytic leukemia secondary to the immunological abnormality caused by the disease or the drugs used in treatment, and specific lesions due to infiltration of the skin by malignant cells (leukemia cutis).[1]

Leukemia cutis represents less than 10% of the skin manifestations of B-cell chronic lymphocytic leukemia and is characterized by solitary, grouped, or widespread papules, plaques, or erythematous tumors that do not ulcerate.[1] These lesions tend to develop on the face, especially on the nose and auricle of the ears (giving rise to the so-called leonine facies), on herpes scars, or in peculiar areas such as the ear lobe, scrotum, or nipple (leukemia lymphatica mamillae).[1] Other clinical presentations are much rarer and include erythroderma, chronic paronychia, subungual nodules with bone destruction, and a papulovesicular eruption on the face.[2]

Oral mucosal lesions in patients with B-cell chronic lymphocytic leukemia are rare and generally occur in the context of nonspecific manifestations of the disease: gingival hyperplasia, petechial hemorrhages, infections, ulcers, and necrosis. Neoplastic lymphocytes seldom infiltrate the palate, with only 4 cases being described in the literature.[3-6]

The patient was a 60-year-old man with a 10-year history of a chronic lymphoproliferative disorder diagnosed as B-cell chronic lymphocytic leukemia. The disease was in an advanced stage and was refractory to alkylating agents. Partial remission was achieved with fludarabine, cyclophosphamide, and mitoxantrone, but treatment was suspended due to hematologic toxicity. The use of