LETTER TO THE EDITOR

ENCEPHALOCRANIOCUTANEOUS LIPOMATOSIS (HABERLAND'S SYNDROME) - A CASE REPORT OF A NEUROCUTANEOUS SYNDROME AND A REVIEW OF THE LITERATURE

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INTRODUCTION

Encephalocraniocutaneous Lipomatosis (ECCL) is a rare neurocutaneous syndrome first described by Haberland and Perou in 1970.[1] It is characterized by unilateral cutaneous, ocular, and neurologic malformations. There are 53 cases described in the literature, but only four of these are in Brazil.[2-10]

CASE REPORT

We present the case of a full-term girl born to non-consanguineous parents. Physical examination revealed a large (14 x 6 centimeters) hairless lesion on the right frontal-parietal scalp (Fig.1), multiple nodular lesions of the right upper eyelid and eyebrow, and a reddish bulbar conjunctival lesion on the right eye consistent with choriocystoma (Fig.2).

A CT scan of the brain showed cranial asymmetry, an arachnoid cyst of the right middle fossa, a right frontal subdural collection, a porencephalic cyst, hemiatrophy of the right hemisphere, and cortical calcifications of the parietal and occipital lobes (Fig.3).

At the age of two years, the patient underwent a complete excision of the scalp lesion after tissue expansion and excision of the nodules on the eyelid (Fig.4). The histopathological examination of the scalp lesion showed absence of hair follicles, a thinned dermis, and extended adipose tissue into the dermis; the cutaneous nodules were

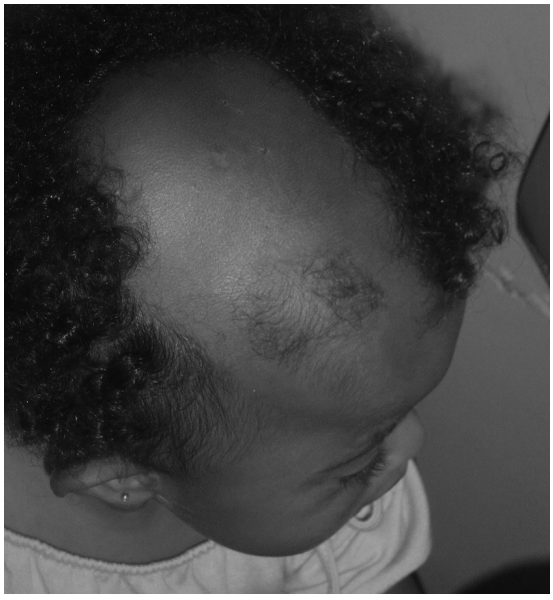

Figure 1 - The hairless lesion ("naevus psiloliparus")

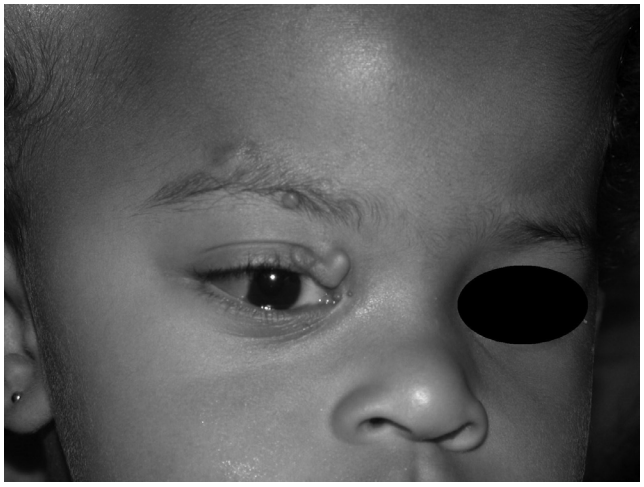

Figure 2 - The choriocystoma and the lipomas of the eyelid

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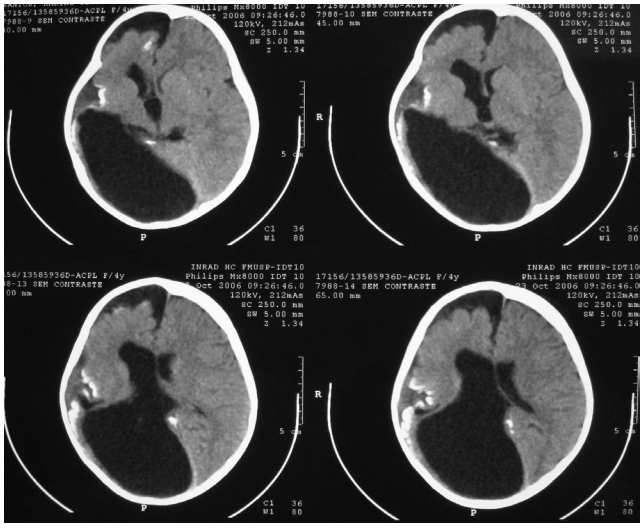

Figure 3 - CT scan of the brain shows: porencephalic cyst, hemiatrophy of the right hemisphere, subdural collection and parietal calcifications

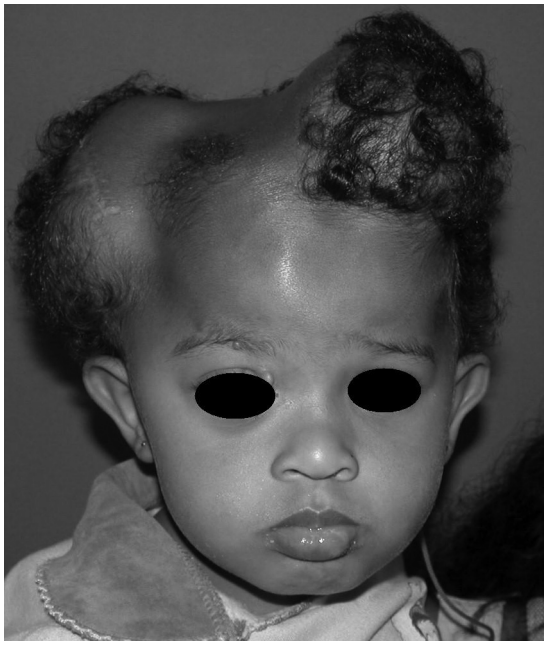

Figure 4 - Tissue expansion for surgical excision of the "naevus psiloliparus"

consistent with lipoma. In the four year follow-up period, the patient was noted to have mental retardation and seizures that were controlled with medication and neurological treatment (Fig.5).

DISCUSSION

ECCL is a rare, sporadic, neurocutaneous syndrome with no predominant gender, racial, or geographical association.[11,12] The genetic mechanism has been hypothesized to involve lethal autosomal dominant genes

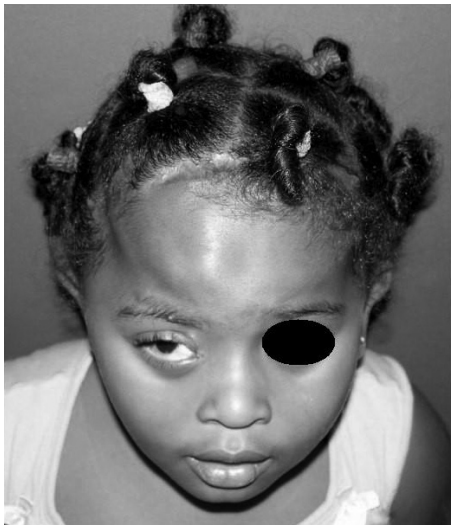

Figure 5 - Post-operative results

that survive by mosaicism, and the pathogenesis is most likely a dysgenesis of the cephalic neural crest and anterior neural tube.[3,11,13]

Clinically, ECCL is characterized by unilateral abnormalities of the brain, eyes, and skin. The most common neurological findings are hemiatrophy, dilated ventricles, porencephalic cysts, abnormal calcifications, intracranial lipoma, and cranial asymmetry. Most patients present with seizures and mental retardation.[3,9,11,12] The hairless lesion of the scalp ("naevus psiloliparus") is pathognomonic, and the papular lesions of the eyelid, consistent with lipomas, are the most frequent feature in all cases reported. However, other lesions have also been reported, such as lipomas of the vertebral spine, odontom, and "café-au-lait" spots.[3,14]

The diagnosis of ECCL may be difficult because of the overlapping features with other neurocutaneous syndromes. Patients with oculocerebrocutaneous syndrome (Delleman's syndrome) have similar lesions of the eyes and scalp, but also present with a pathognomonic mid-hindbrain malformation. Other syndromes, like Sturge-Webber syndrome and Proteus syndrome, have neurological features similar to ECCL, although the cutaneous malformations are very different.[3,6,11,14] Therefore, a careful and complete examination of these patients including a multidisciplinary evaluation and follow-up by a neurosurgeon, pediatrician, ophthalmologist, and plastic surgeon is necessary for accurate diagnosis of this condition,. Delay in diagnosis is common in these patients though it is best if the pediatrician makes the diagnosis at birth to improve prognosis and orientations of the relatives. Antenatal diagnosis is not usually made because the intracranial malformations noted on an antenatal sonogram are not specific for ECCL.[15]

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