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A Filipino male with encephalocraniocutaneous lipomatosis (Haberland's syndrome)

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Abstract

Background: Encephalocraniocutaneous lipomatosis (ECCL), also known as Haberland's Syndrome, is a sporadically occurring neurocutaneous syndrome with no gender or race predilection. ECCL patients present with a broad spectrum of clinical manifestations, often in a unilateral distribution. The hallmark of ECCL is the nevus psiloliparus, a soft, bulging, lipomatous scalp lesion, with associated alopecia.

Main observations: We describe a case of a 2-month-old Filipino male with a soft, ill-defined mass with associated alopecia on the fronto-parietal scalp. Biopsy revealed findings consistent with a nevus psiloliparus. The patient also presented with a lipomatous nodule on the right temple, as well as choristomas and a coloboma on the right eye. He had no history of seizures and development was at par with age.

Conclusion: Recognition of ECCL is important in order to work-up the patient for concomitant problems, such as central nervous system and cardiac anomalies, and employ a multidisciplinary approach in the management of these patients. (*J Dermatol Case Rep*. 2013; 7(2): 46-48)

Introduction

Encephalocraniocutaneous lipomatosis (ECCL) was first described by Haberland and Perou in 1970. It is a sporadically occurring neurocutaneous syndrome with no gender or race predilection. ECCL patients present with a broad spectrum of clinical manifestations, often in a unilateral distribution.[1,2] The hallmark of ECCL is the nevus psiloliparus, a soft, bulging, lipomatous scalp lesion, with associated alopecia. A study conducted by Moog in 2009, identified 54 cases of ECCL.[1] Review of these cases, PubMed and the Health Research and Development Network (HERDIN) revealed no reported cases of ECCL in a Filipino.

Case Report

We present the case of a 2-month-old Filipino male from Manila, Philippines, who was born full-term via spontaneous vaginal delivery to non-consanguineous parents. There was no known maternal exposure to teratogens. The patient had good cry and activity at birth and had no episodes of seizures or cyanosis. His developmental history was at par with age. His family medical history was unremarkable.

Systemic physical and neurologic examination revealed essentially normal findings. Dermatologic examination revealed a soft, ill-defined mass with an overlying smooth alopecic patch on the right parieto-occipital scalp (Fig. 1). A soft, moveable, nontender, nonerythematous nodule was noted on right temple. Smooth fleshy masses were noted from the lateral and medial canthi of the right eye, extending to the corneal surface, consistent with a choristoma. Further ophthalmologic examination revealed an optic disk coloboma on the right eye.

Skull x-ray revealed an intact cranial vault and no abnormal intracranial calcifications. Cranial ultrasound showed no intracranial extension of the scalp lesion. Brain parenchyma was unremarkable with normal echogenicity. No mass lesions, abnormal calcifications or extra-axial fluid collection were observed. Punch biopsy of the scalp revealed thick subcutaneous fat in the superficial reticular dermis (Fig. 2). Punch biopsy of the mass on the right temple revealed pockets of adipocytes within the dermis surrounded by collagen bundles with areas of fibrosis.

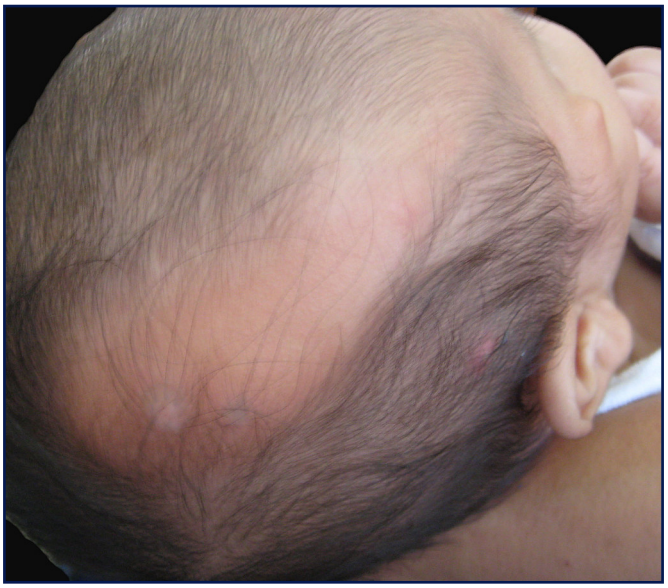

Figure 1

Soft, ill-defined mass with overlying alopecic patch on the right parieto-occipital scalp.

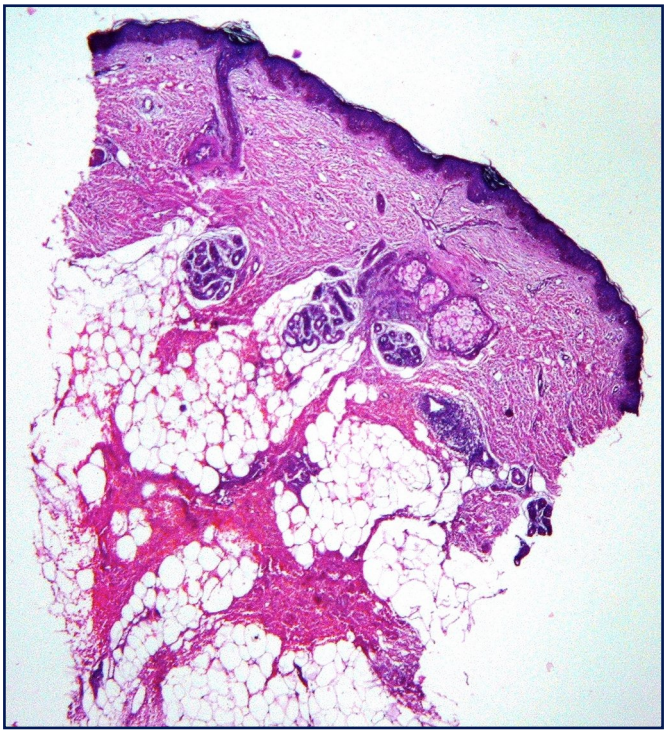

Figure 2

Biopsy of the scalp lesion revealing subcutaneous fat in the reticular dermis.

Discussion

ECCL or Haberland's syndrome is a multisystemic disorder affecting the skin, eyes and central nervous system. Odontogenic and cardiac findings have also been described. The characteristic cutaneous finding is the nevus psilolipa-rus (hairless fatty tissue nevus). Histopathology of this lesion shows focal dermal fibrosis with subcutaneous fat in the reticular dermis. Other skin findings that may be present include lipomatous nodules on the face and eyelid. These are often ipsilateral to the nevus psiloliparus.[3,4] The most common ophthalmologic finding is a choristoma, such as an epibulbar and limbal dermoid or lipodermoid. ECCL patients may also present with colobomas, corneal and scleral abnormalities, aniridia, microphthalmia and globe calcification.[1]

Clinically, ECCL patients present with a wide range of neurological manifestations — from having normal development without seizures to having intractable seizures and severe mental and psychomotor delay. Imaging studies reveal variable brain and spinal anomalies, including lipomas, calcifications, hemispheric atrophy, and hydrocephalus. Nonetheless, it has been noted that the severity of the neurologic lesions do not correlate with the severity of neurologic manifestations or the degree of skin and ocular lesions.[1]

Skeletal findings that have been described among ECCL patients include jaw tumors and multiple bone cysts. Congenital heart defects have also been observed in some patients.[1,5]

Diagnostic Criteria

Based on a review of 54 cases, Moog formulated diagnostic criteria for ECCL.[1] Our patient fulfilled one major skin criterion (proven nevus psiloliparus) and one minor skin criterion (subcutaneous lipoma on the frontotemporal region). He also had a choristoma (major eye criterion) and a coloboma (minor criterion). A definite case was defined as having 2 systems involved with major criteria one of which is a proven nevus psiloliparus, as in our patient.

Differential Diagnosis

Oculoectodermal syndrome is a condition characterized by choristomas, particularly epibulbar dermoids, and non-scarring alopecia. The typical scalp lesion however is aplasia cutis congenita. Facial and intracranial lipomas are unusual.[1,6]

Proteus syndrome is a sporadic multisystemic condition that may also present with cutaneous lipomas and ocular choristomas in a mosaic distribution. However, this condition is characterized by progressive, asymmetric, disproportionate overgrowth, especially of the limbs, skull, external auditory meatus, and viscera. The characteristic skin lesion is a cerebriform, connective tissue nevus. Patients with Proteus syndrome may also present with multiple epidermal nevi, lipomas or regional lipohypoplasia and vascular malformations.[7]

Oculocerebrocutaneous syndrome (OCCS), also known as Delleman syndrome, is also characterized by congenital skin, eye and brain anomalies. Lesions are also often unilateral. It is distinguished from ECCL by the most common dermatologic findings being a crescent-shaped suprauicular aplasia or hypoplasia, and a striated muscle hamartoma.[1,8,9]

Pathogenesis

The pathogenesis of ECCL is still unknown. It has been hypothesized to be a mesenchymal disorder affecting neural crest derivatives. In 1993, Happle suggested that ECCL

may be due to a lethal somatic mutation surviving in the mosaic state. According to Happle's theory, certain postzygotic mutations occur that would otherwise lead to early death of the embryo. These cells however, manage to survive in a mosaic state, lying in close proximity to normal cells. Individuals with these mutations would be unable to transmit the mutation to their offspring as this would lead to death in-utero.[10] This theory is consistent with the sporadic nature of the disease and the variable severity of manifestations.

Diagnosis and Treatment

Diagnostic examinations for ECCL patients include a biopsy of skin lesions and an ophthalmologic exam. Neuroimaging studies should be done to assess for central nervous system anomalies. An ECG and a 2D-echo may help screen for cardiac problems.

Treatment for ECCL is symptomatic. Surgery may be done for the cutaneous lesions. Antiepileptic treatment for seizures, and anti-arrhythmics may be given if necessary for cardiac symptoms.

Conclusion

In conclusion, we have presented a 2-month-old Filipino male with multiple right-sided lesions on the scalp, temple and eye. The patient was diagnosed with ECCL or Haberland's syndrome. Recognition of this rare syndrome is important in order to work-up the patient for concomitant problems, such as CNS and cardiac anomalies, and employ a multidisciplinary approach in the management of these patients. Also, knowing the sporadic nature of this disease will help guide genetic counseling.

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