Encephalocraniocutaneous lipomatosis (Haberland syndrome): A rare case report

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ABSTRACT

Haberland syndrome or Fishman syndrome also known as encephalocraniocutaneous lipomatosis (ECCL) is a rare, congenital neurocutaneous disorder. It is characterized by unilateral involvement of skin, eyes and central nervous system. We report the case of a 28-year-old woman who presented with soft lipomatous swelling over right temporal area with nonscarring alopecia of part of frontal and parietal region. The patient had a history of seizures and ipsilateral scleral dermoid. Computed tomography scan findings were suggestive of lipomas and calcification of falx. Magnetic resonance imaging showed right-sided hemi atrophy and two intracranial cysts. We report this case because of its rarity.

Key words: Encephalocraniocutaneous lipomatosis, Fishman syndrome, Haberland syndrome, lipomatosis

INTRODUCTION

Haberland syndrome, or encephalocraniocutaneous lipomatosis (ECCL), is a rare neurocutaneous syndrome reported since 1970 by Haberland and Perou.[1] It was described as a new example of ectomesodermal dysgenesis characterized by mental retardation; early onset of seizures; unilateral, temporofrontal lipomatosis; ipsilateral, cerebral, and leptomeningeal lipomatosis; cerebral malformation and calcification; and lipomas of the skull, eye, and heart.

CASE REPORT

A 28-year-old female patient having a history of seizures and eye abnormality was referred to dermatology department for evaluation of skin lesions over right temporal region and scalp. The skin lesions were nonitchy, nontender, and not increasing in size. On examination single, soft, oval nodule was found over right temporal region. There was a single linear plaque extending from scalp to eyebrow [Figure 1], single soft nodular lesion over right eyebrow and few papules over right eyelid [Figure 2]. Clinical impression of lesion was of nevus psiloliparus. Ophthalmological examination showed dermoid tumor of scleral limbus [Figure 2], conjuctival tumor and small papule around eyelid, and optic pallor on fundoscopy.

Histopathology revealed lobules of diffusely arranged mature adipocytes separated by congested capillaries [Figure 3]. There was right-sided frontal and parietal prominence and nonscarring soft area of hairloss. A plain non contrast computed tomography of brain showed fat density along falx cerebri and extensive calcification along right cerebral cortex convexity with right cerebral hemiatrophy. Magnetic resonance imaging of brain revealed right-sided cerebral hemiatrophy, abnormal leptomeningeal enhancement in cortical sulci of right temporo-occipital region as well as dura over right temporo-occipital convexity, two intracranial cysts—one in right temporal lobe and second in velum interpositum [Figure 4] and

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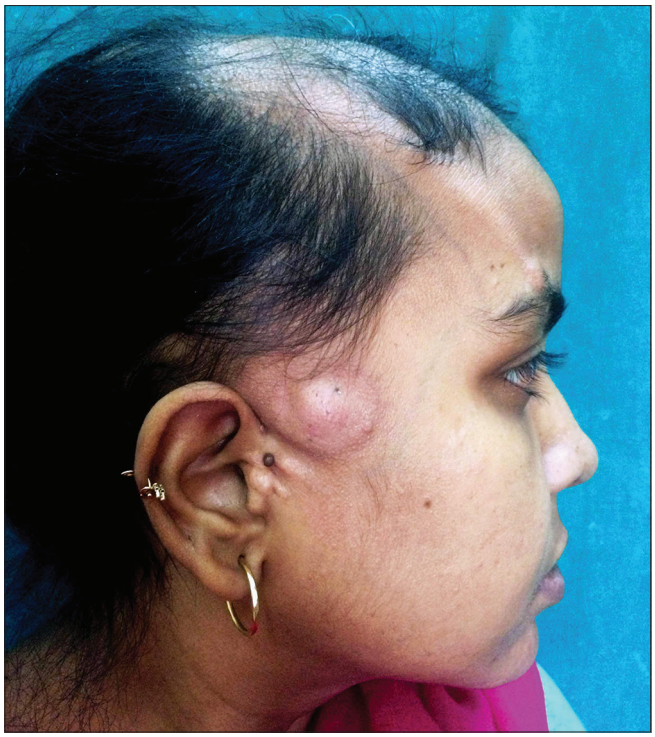

Figure 1: Single linear plaque extending from scalp to eyebrow, clinically suggestive of lipomatosis

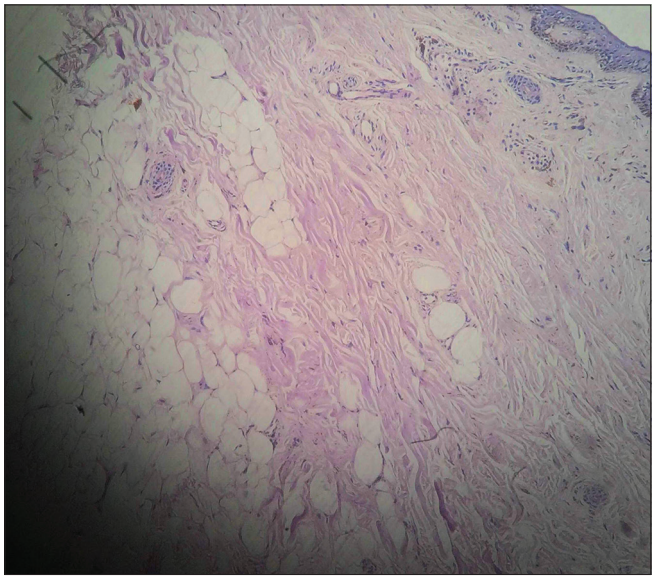

Figure 3: Histopathology: Lobules of diffusely arranged mature adipocytes separated by congested capillaries (H and E, ×10)

falx-based calcification and lipomas, which were more evident on computed tomography.

The patient was born out of non-consanguineous marriage. Delivery was normal, spontaneous, and without any complications. Behavioral observations revealed a normal mental status. Electrocardiogram, chest radiography, complete blood count and the biochemistry panel were within normal

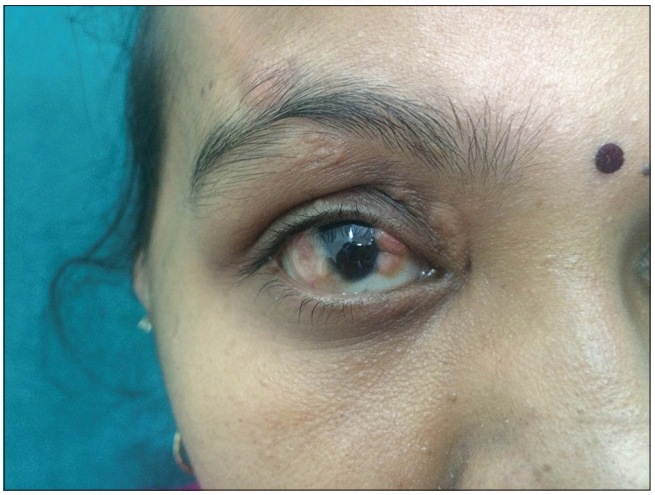

Figure 2: Dermoid tumor of sclera limb, single soft nodular lesion over right eyebrow and few papules over right eyelid

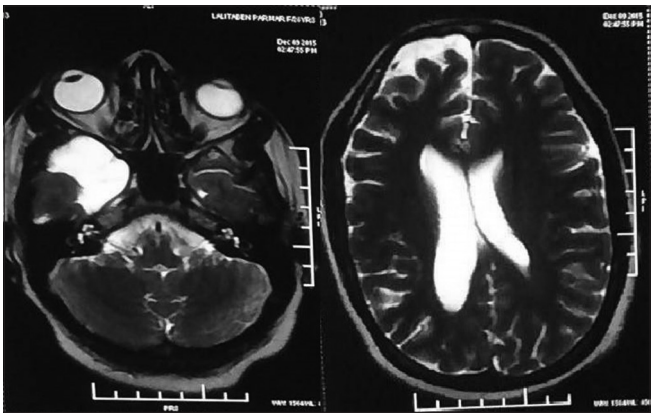

Figure 4: Magnetic resonance imaging showing cyst in right temporal lobe and in velum interpositum and dilated lateral ventricle

limits. Further enquiry revealed history of epilepsy, which was of generalized tonic–clonic type since 3 years. According to revised criteria described by Moog in 2009 we put the diagnosis of Haberland syndrome.[2]

DISCUSSION

ECCL is a rare neurocutaneous syndrome. The first case was reported by Haberland and Perou in 1970.[1] Subsequently Fishman *et al*. reported additional cases.[3] It is characterized by congenital, unilateral cerebral malformations with ipsilateral scalp, face, and eye lesions. It is mainly unilateral, but bilateral cases have also been reported.[4] Several primary clinical features had been described including seizure, development delay, mental retardation, or spasticity of the contralateral limbs, which were most related to the intracranial components. Haberland and Perou first described ECCL in 1970. Diagnosis of ECCL is made by clinical presentation, image studies, and histopathological findings. As ECCL stands for encephalocraniocutaneous lipomatosis, almost all cases

show typical cutaneous and central nervous system (CNS) abnormalities. The pathogenesis and molecular basis of ECCL remains unknown to date. There is no racial or gender predilection. The widely accepted hypothesis for this congenital anomaly is somatic mosaicism caused by lethal autosomal mutations, which survives only in a mosaic state.[5] Several diseases such as Proteus syndrome, Delleman syndrome, and neurocutaneous melanosis are supposed to originate from similar cutaneous mosaicism of lethal mutations as in ECCL. All patients with ECCL should undergo detailed neuroimaging studies since the neurological manifestations can vary from being totally normal to severe vascular malformations. Most common neurological deficits include mental retardation, spasticity, facial palsy, hemiparesis or hemiplegia, and epilepsy.[6] Cases of mild ECCL might have been underdiagnosed or misdiagnosed. A special finding, named nevus psiloliparus by Happle and Kuster for description of hairless fatty nevus, explained the regional alopecia due to the underlying scalp lipomatosis. Nevus psiloliparus is a frequent finding in ECCL rather than a diagnostic criterion. Clinically main differential diagnosis includes proteus syndrome, neurofibromatosis, Sturge–Weber syndrome, epidermal nevus syndrome, Goldenhar syndrome.[7] The presence of certain distinct cutaneous and CNS features associated with alopecia, distinguish all the above-mentioned disorders from ECCL. Treatments for ECCL are mainly symptomatic. Cutaneous or ocular lipomatous hamartoma can be removed surgically. Patients with seizure disorder will benefit with antiepileptics. Neurosurgery or other invasive procedures should be spared for the cases refractory to medical treatments. The prognosis of ECCL is considered to be directly related to the extent of neurological disease. To conclude, due to rarity of this syndrome and involvement of multiple systems, a multidisciplinary approach helps to clinch the diagnosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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