Case Report

Partial Encephalocraniocutaneous Lipomatosis Syndrome

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Encephalocranial lipomatosis is a rare disorder that characteristically involves ectomesodermal tissues such as skin, eye, and the central nervous system. Here, we report a 3-year-old girl presented with developmental delay, seizures, limbal dermoid, and weakness of right lower limb. Imaging revealed hemiatrophy, arachnoid cyst, and polymicrogyria. The constellation of clinical finding and imaging leads to the diagnosis.

Keywords: Encephalocranial lipomatosis, microcephaly, ocular dermoid

INTRODUCTION

Encephalocraniocutaneous lipomatosis (ECCL) is a rare neurocutaneous syndrome first described by Haberland and Perou in 1970.[1,2] So far, 53 cases have been described in the literature. It is characterized by unilateral lipomas of the cranium, face, and neck, ipsilateral lipodermoids of the eye, and ipsilateral brain anomalies.[3] Here, we report a female child who presented to our institute with the peculiar findings.

CASE REPORT

A 3-year-old female child born to third-degree consanguineous parents with normal birth history was admitted for a brief generalized tonic–clonic seizure episode. She had a similar episode at 7 months and 2½ years of age in the past. The child had mild motor and language developmental delay (started walking at 1½ years, started speaking at 2 years of age). Family history was negative. One of her elder brothers was normal.

On examination

The child had microcephaly (head circumference: 44.5 cm, <3rd percentile), and she was able to interact with the family members. Development quotient was 97, and she had mild facial atrophy on the left side and dermolipoma on superotemporal aspect of the limbus in the left eye [Figure 1]. Fundus examination revealed medullated nerve fiber. She had subtle weakness in her right lower limb. Rest of the neurological examination was normal. Cardiac and ENT evaluation were normal. Ultrasound sonography abdomen,[4,5] X-ray spine, renal function test, and liver function test were normal. Magnetic resonance imaging brain showed atrophy of left hemisphere [Figures 2 and 3] with left middle cranial fossa arachnoid cyst [Figure 4] and left tempooccipital polymicrogyria [Figure 5]. Electroencephalogram showed evidence of mild dysfunction on the left side.

DISCUSSION

Underlying defect in encephalocranial lipomatosis syndrome has been speculated to result from action of lethal autosomal dominant gene that survives in...
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...the mosaic state. (The pathogenesis of the syndrome still remains unclear. Till date, there is no evidence of genetic transmission or chromosomal abnormality. Somatic mosaicism is thought to be the underlying pathophysiology in ECCL.)[6] It usually presents with porencephalic cyst with cortical atrophy, cranial asymmetry, developmental delay, seizures, ipsilateral lipomatous hamartomas of the scalp, eyelid, and spasticity of contralateral limbs. Some cases show bilateral involvement.[7] Other diseases that superficially resemble ECCL and are often associated with neurological abnormalities are epidermal nevus syndrome, focal dermal hypoplasia, Proteus syndrome, and Goldenhar syndrome. Although epibulbar choristomas and limbal dermoids can occur sporadically in isolation or in a Mendelian inheritance pattern, there may be systemic associations such as Goldenhar’s syndrome, the linear nevus sebaceous syndrome, or ECCL. (Complications related to the central nervous system malformation may be a cause of increased morbidity and mortality in these patients. These patients are also at risk for developing certain neoplasms such as juvenile extranasopharyngeal angiofibroma of gingiva, papillary glioneuronal tumor, and low-grade glioma/astrocytoma.)[8,9] In our case, the child had dermolipoma in the left eye, facial atrophy on the left side, microcephaly, hemiatrophy of brain, arachnoid cyst and polymicrogyria in the left side, seizure episode (about two-thirds of patients have a normal development or mild mental retardation and half have seizures[7,10]), and mild developmental delay. As there is no cutaneous manifestation in our case, it is incomplete syndrome of ECCL.

**CONCLUSION**

Case is reported here for its rarity. Limbal dermolipoma may be an external marker of underlying brain anomaly. In a child with limbal dermolipoma, developmental delay, or seizures, one has to screen for brain anomaly.

**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

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**Figure 2:** Magnetic resonance imaging brain showing left hemiatrophy T2 image

**Figure 3:** Magnetic resonance imaging brain showing left hemiatrophy coronal image

**Figure 4:** Magnetic resonance imaging showing arachnoid cyst

**Figure 5:** Magnetic resonance imaging of brain showing occipital polymicrogyria
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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