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## Case Report

# Encephalocraniocutaneous lipomatosis: A rare congenital neurocutaneous syndrome

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### ABSTRACT

Encephalocraniocutaneous lipomatosis (ECCL) is a rare sporadic congenital neurocutaneous disorder with quite specific clinical features and neuroimaging pattern that is well seen on MR imaging.

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## Introduction

Encephalocraniocutaneous lipomatosis (ECCL), also known as Haberland syndrome is a rare sporadic congenital neurocutaneous disorder characterized by unilateral lipomas of the cranium, face and neck, ipsilateral lipodermoids of the eye and ipsilateral brain anomalies. ECCL syndrome has quite a specific neuroimaging pattern that is well seen with MR imaging.

## Case report

A 2-year-old child who presented with global developmental delay and seizures since 5 months of age along with a non-progressive swelling in left eye since birth. Antenatal history was uneventful and the child was delivered by normal vaginal route at term with normal cry at birth. On physical examination, a patch of alopecia was noted on scalp on left side along with epibulbar dermoid in the left eye [Figure a and b].

A preliminary noncontrast CT scan of head was done in the emergency radiology department following an episode of generalized tonic-clonic seizures that showed atrophy of the

left cerebral parenchyma with prominent ventricular system more pronounced in the occipital horn of left lateral ventricle (Fig. c) along with few intracranial lipomas which were noted at prepontine cistern and cerebellopontine angle on left side.

The child was further evaluated with MRI. MRI brain on a 3T MRI scanner demonstrated multiple left cerebral abnormalities including cerebral atrophy, paucity of temporoparietal white matter and an abnormal cerebral cortical morphology (polymicrogyria) (Fig. 1g-i). Few extra-axial lipomas in the prepontine cistern and at the cervicomedullary junction were also noted on T1W MR images. Few extra-axial lipomas were also noted at the cervicomedullary junction (Fig. 1k).

## Discussion

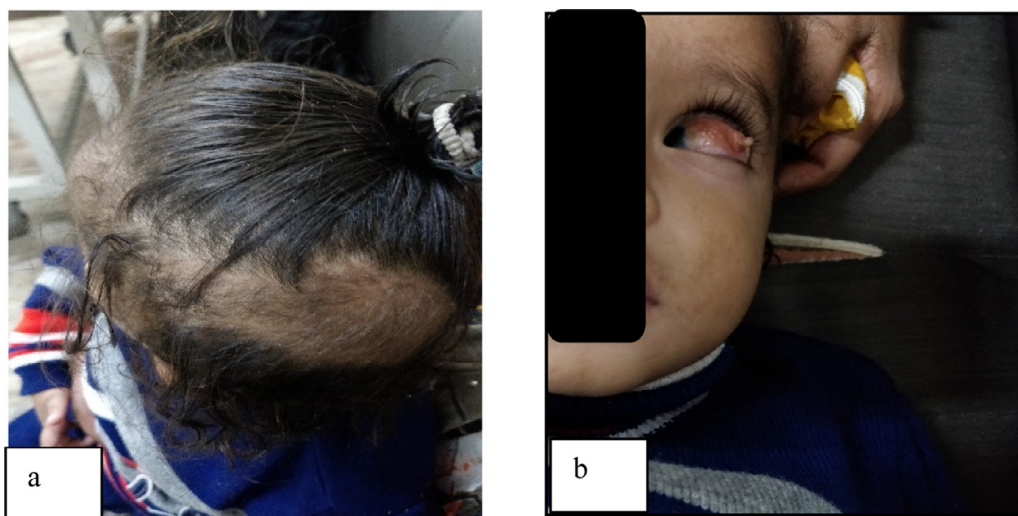
Neurocutaneous syndromes/phakomatoses represent a group of central nervous system disorders with concurrent lesions in skin, eye, and other visceral organs. ECCL (also called Fishman or Haberland syndrome) is a rare congenital but nonhereditary neurocutaneous syndrome characterized by unilateral skin, facial, and ocular lesions and ipsilateral cerebral malformations. Fewer than 54 cases have been reported since 1970. It

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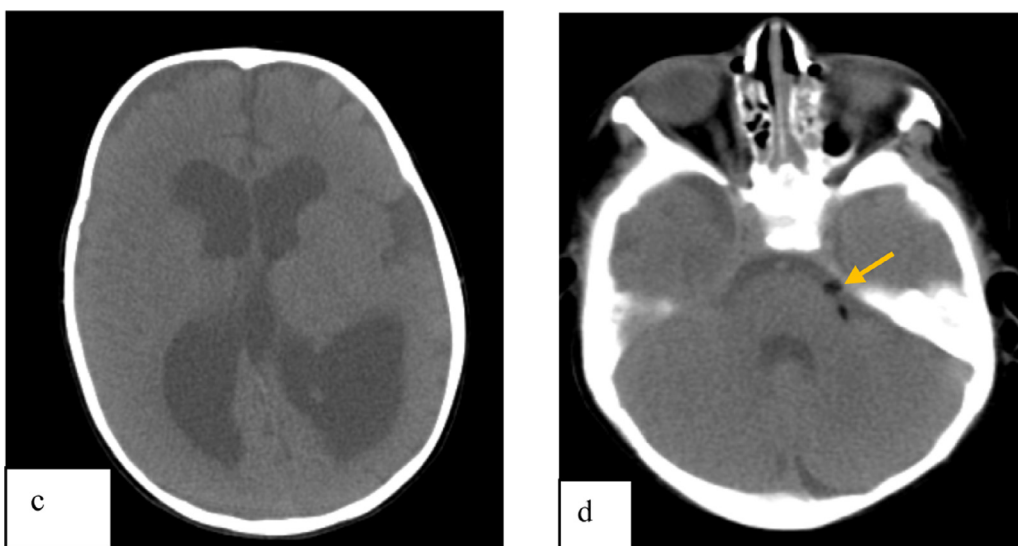
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**Fig. 1(a & b) – Figure a & b** showing patch of alopecia on the scalp and epibulbar dermoid on left side. Showing a patch of alopecia on scalp on left side along with epibulbar dermoid in the left eye.



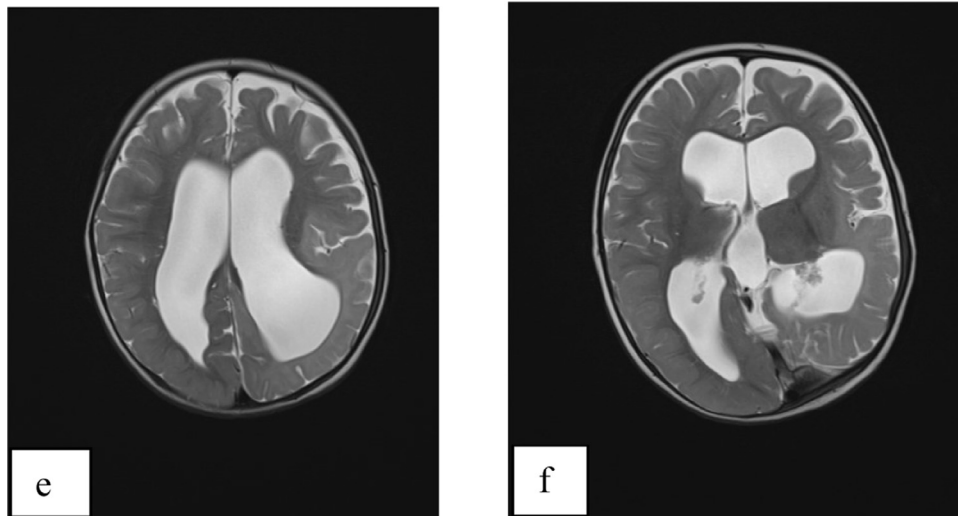
**Fig. 1(c and d) – Figure c and d:** Non contrast CT images of the child showing dilated ventricular system most pronounced in the occipital horn of left lateral ventricle along with few intracranial lipomas (arrow).

occurs due to sporadic mutations in the *FGFR1* gene which is involved in making a protein called fibroblast growth factor receptor that triggers signaling within cells important for the normal development and growth of several parts of the body, including the brain [1–3].

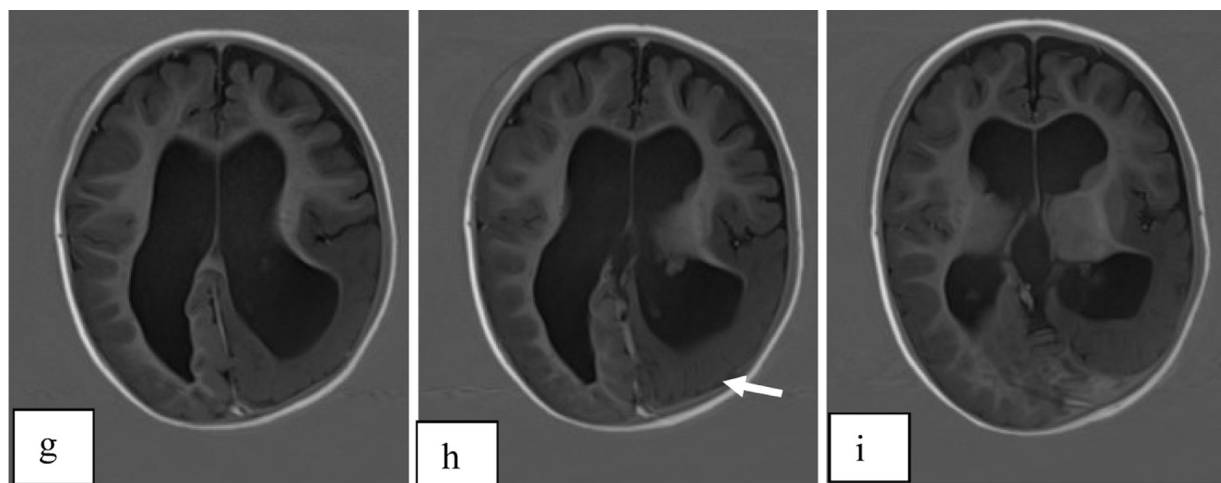
These children usually present with marked developmental delay, mental retardation, seizures, spasticity of the limbs along with characteristics lipomatous hamartomas of scalp, eyelids, outer globe of the eye, patchy alopecia, etc., which are present since birth and are nonprogressive in nature. Our patient had a patch of alopecia and an epibulbar dermoid in left eye since birth along with left cerebral abnormalities. Other kinds of hamartomatous conditions have also been reported including noncancerous jaw tumors, which however were not seen in our patient.

The primary imaging features include cerebral hemispheric atrophy ipsilateral to scalp and eye lesion, enlargement of ipsilateral lateral ventricle, intracranial, and intraspinal lipomas, abnormal cerebral cortical morphology such as polymicrogyria in temporal, parietal, and occipital lobes, all of which were noted in our patient. Presence of intracranial cysts, for example, porencephalic cysts, arachnoid cysts, gyral calcifications, and abnormal diffuse ipsilateral leptomeningeal enhancement has also been reported in some patients previously [4].

Treatment of this condition is usually supportive and includes treatment of seizures. Ophthalmic management is limited to excision of choristomas with or without lamellar keratoplasty, removal of cutaneous lesions for cosmetic improvement, and low vision rehabilitation. Prevention by antenatal



**Fig. 1(e and f) – Figure e and f:** T2W axial MR images showing left cerebral hemiatrophy along with marked paucity of white matter in the left cerebral parenchyma most marked in left parietal lobe.



**Fig. 1 (g-i) – Figure g-i:** Inversion recovery axial MR images showing left cerebral hemiatrophy along with marked paucity of white matter in the left parietal lobe. Multiple small and broad gyral convolutions separated by shallow sulcal spaces can be seen in left temporo-occipital lobe reaching up to perisylvian region suggestive of polymicrogyria.

diagnosis is usually not possible since intracranial malformations on antenatal sonogram are nonspecific [5].

A constellation of specific clinical and radiological findings can correctly diagnose this rare neurocutaneous syndrome. Our patient had abnormal cerebral cortical morphology, intracranial lipomas, cerebral hemiatrophy ipsilateral to the skin and eye lesion, all pointing toward the diagnosis of ECCL.

### Why is there a need to diagnose this condition accurately?

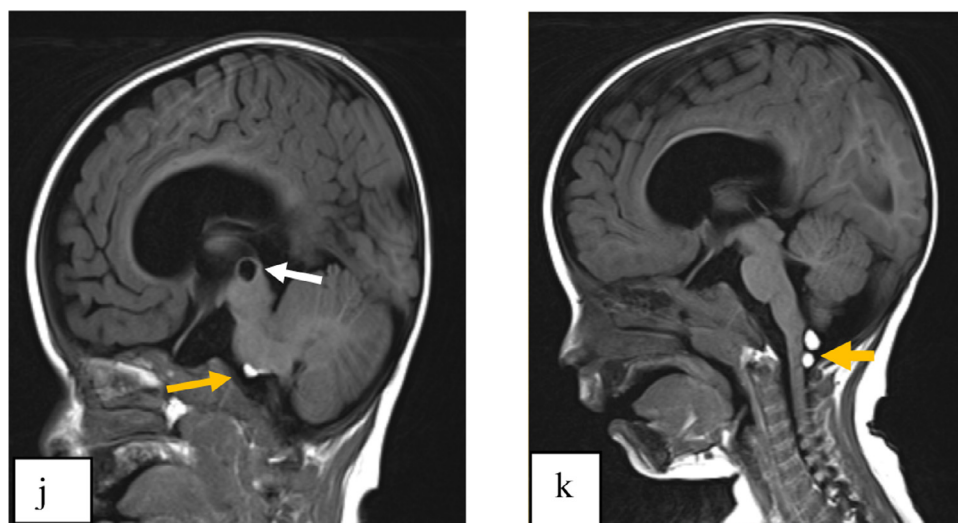
Unlike most of the neurocutaneous syndromes which are inherited in an autosomal dominant fashion, ECCL is due to sporadic mutation. So if the parents are getting apprehensive

about similar occurrence in their next child, they can be counselled that this syndrome is nonhereditary. Complications related to central nervous system (CNS) malformation cause morbidity and mortality due to risk for neoplastic conditions like papillary glioneuronal tumor and low-grade glioma/astrocytoma. Therefore, screening for these conditions is critical during follow-up of an asymptomatic child [6,7].

### Differential diagnosis

- Proteus syndrome:

This is a rare multisystemic hamartomatous condition characterized by asymmetrical overgrowth of almost any part of the body. In this syndrome, affected patients are normal



**Fig. 1(j and k) – Figure j and k: T1W sagittal MR images showing hyperintense lipomas in the prepontine cistern and at the cervicomedullary junction (yellow arrows). Also noted was a small cystic lesion (1.2 cm size) along posterior wall of III ventricle in midline following cerebrospinal fluid (CSF) signal intensity, likely an ependymal cyst (white arrow).**

at the time of birth and they progressively develop lesions in childhood unlike ECCL where cutaneous and ocular lesions are present since birth and are static in course. Hemimegalencephaly is the prominent CNS manifestation in this syndrome. The characteristic intracranial lipomas of ECCL are not found in this syndrome.

- **Sturge-Weber syndrome:**

There is also a superficial resemblance of ECCL to Sturge-Weber syndrome (SWS). In both the conditions, hemiatrophy and gyriform parenchymal calcifications can be seen. The gyriform calcifications are detectable in the first few months of life in patients with ECCL in contrast to SWS where they are seldom evident before 1 year of age. Also, the characteristic cerebral lipomas documented in ECCL do not occur in patients with SWS.

- **Sebaceous nevus syndrome**

Choristomas, epilepsy, and mental retardation also occur in sebaceous nevus syndrome, with the cutaneous epidermal nevus itself affecting the face and upper part of the trunk and frequently are located in the midline. Eyelid involvement by the sebaceous nevus and epibulbar choristomas are the most common ocular manifestations. Some authors believe that ECCL and sebaceous nevus syndrome may represent a continuum of phenotypic expression [8].

- **Oculocerebrocutaneous syndrome/Delleman syndrome**

In this syndrome, there are characteristic orbital cysts, microphthalmia, and aplastic skin defects in a patchy arrangement with absence of facial lipomas and scalp alopecia.

Additionally, CNS malformations are rare in the syndrome and limited to intracranial cysts and agenesis of corpus callosum [8].

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