

Encephalocraniocutaneous lipomatosis: A rare neurocutaneous syndrome

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ABSTRACT

Encephalocraniocutaneous lipomatosis is a congenital hamartomatous disorder with unique ocular, cutaneous and neurological features. A 13-year-old boy presented with history of mental retardation and delayed developmental milestones. Bulbar conjunctiva of left eye showed hypertrophy with a soft reddish limbal nodule encroaching on the cornea. Dermatological examination showed multiple patches of alopecia, soft papules in the left perioral and periorbital areas, soft masses over the right axilla, trunk and in the lumbosacral region suggestive of lipomas. The CT scan of the brain revealed well-defined, hypodense lesions in both the cerebellar hemispheres suggestive of lipomas. The constellation of these findings led us to a diagnosis of encephalocraniocutaneous lipomatosis.

Key Words: Cutaneous, Cranial, Lipomas

INTRODUCTION

Encephalocraniocutaneous lipomatosis (ECCL) is an uncommon neurocutaneous syndrome, first described by Haberland and Perou in 1970.^[1] This condition is also known as Fishman syndrome.^[2] It is characterized by unilateral eye lesions in the form of choristomas and skin lesions like lipomas, connective tissue nevi and alopecia, in association with ipsilateral cerebral malformations and porencephalic cysts, seizures, mental retardation. Visceral lipomas, nevus sebaceous and other organoid nevi can also be seen.^[1,2] Since its first description in 1970, not more than 20 cases have been reported in the literature. To the best of our knowledge, this is probably the first case of its kind to be reported from the Indian subcontinent.

CASE REPORT

A 13-year-old boy was admitted with complaints of fever, vomiting and convulsions since ten days. He had a history of a

ventriculo-peritoneal shunt done at the age of six months which was removed after a year due to infection. He had a history of mental retardation, delayed developmental milestones, mass in left eye, localized patches of alopecia on the scalp and frequent admissions.

The patient's general examination revealed normal vital parameters. He was conscious, irritable and moving all four limbs. No neck stiffness or papilloedema were found. Cardiovascular and respiratory systems were within normal limits. Epigastric hernia was noted on per abdominal examination. Bulbar conjunctiva of left eye showed hypertrophy with a soft reddish limbal nodule encroaching on the cornea. Genital examination revealed left undescended testis with inguinal hernia and hydrocele of right spermatic cord. Dermatological examination showed multiple patches of smooth, hairless skin with normal markings but absent hair follicles on the scalp, three to four soft skin-colored papules in the left perioral and periorbital areas, soft masses over the

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right axilla and trunk and a transverse soft mass crossing the midline in the lumbosacral region suggestive of lipoma. Teeth, nails and mucosa were normal.

No abnormality could be detected on hematological and cerebrospinal fluid examination. The computerized tomography (CT) scan of the brain revealed well-defined, hypodense lesions in both the cerebellar hemispheres suggestive of lipomas. Scattered areas of calcification were noted in the right occipital and frontoparietal regions. Right porencephalic cyst was observed communicating with and distorting the right lateral ventricle, third ventricle and fourth ventricle. No other systemic abnormality was noted after investigations.

Histopathological examination of superior bulbar conjunctival mass showed adipose tissue in the submucosa, thick walled blood vessels and islands of cartilage typical of choristoma. On biopsy, the soft lumbosacral mass and perioral nodules showed excessive adipose tissue in the dermis suggestive of lipoma while periorbital nodules revealed connective tissue nevi.

DISCUSSION

ECCL or Fishman syndrome is a rare neurocutaneous syndrome of unknown cause. The genetic mechanism underlying these complex birth defects has been hypothesized to result from the action of lethal autosomal dominant genes surviving by mosaicism.^[3,4] Although the exact embryologic defect responsible for ECCL is yet to be determined, the embryonic origin of the segmental choristomas and hamartomas is thought to be neural crest-derived mesectoderm.^[5,6] The hypothesis of the causal role of anomalous neural crest morphogenesis and migration in the development of these abnormalities is supported by a rare case report of neurocutaneous melanosis in association with ECCL.^[7] Several other hypotheses like maternal viral infection during pregnancy, dysgenesis of cephalic neural crest and anterior neural tube have been proposed by previous authors.^[8-10]

Although a spectrum of clinical features exists in the cases of ECCL reported so far, certain neurocutaneous findings appear in almost every patient. The abnormalities tend to be unilateral and only four cases with bilateral involvement have been encountered.^[8,10-12] In our patient, the cerebral parenchymal calcification, porencephalic cyst and hydrocele of spermatic cord were right-sided. The left-sided lesions were bulbar choristomas, connective tissue nevi of skin and eye, undescended testis and inguinal hernia, while alopecia and lipomas were bilateral. In all but two cases in the literature,^[9,11] lipomas and neurocutaneous findings were limited to the head

and central nervous system. However, in our patient, the lipoma was noted crossing the midline in the lumbosacral area similar to the lesion described by Al Mefty *et al.*^[11] Ocular abnormalities are supposed to be the most consistent feature of ECCL and the commonest findings are epibulbar choristomas and small skin nodules around the eyelids.^[5,10] Our patient had epibulbar choristoma in the right eye and bulbar conjunctival hypertrophy in both eyes. Histopathological examination of perioral nodules showed connective tissue nevi while perioral nodules revealed lipomas.

In ECCL, central nervous system (CNS) involvement appears to be variable with mild to severe mental retardation and developmental delay having been present in all but two cases reported in the literature.^[13,14] Seizures have been reported in all but three cases.^[5,12,13] Our patient demonstrated all these manifestations. On radiological examination, porencephalic cyst was the most frequently reported anomaly as mentioned by Kodsí *et al.*^[5] The other abnormalities reported were lateral ventricle cyst,^[7] temporal lobe defect,^[9] dilated right ventricle, corpus callosum agenesis^[12] and CNS lipomatosis. The CT scan studies of our patient exhibited hypodense lesions in both cerebellar hemispheres suggestive of lipoma, porencephalic cyst and distortion of right lateral and third and fourth ventricles. An unusual finding in our patient was cerebellar and cerebral calcification which has been previously reported only by Haberland^[9] in his original patient of ECCL. Shah *et al.*^[14] reported a 30-year-old woman with history of tinnitus and progressive hearing loss on her right side. The magnetic resonance imaging revealed multiple intracranial lipomas with schwannoma at the cerebellopontine angle and multiple lipomas over both upper arms. However, other cerebral and ophthalmic abnormalities were not observed in their patient. Also unique in our patient, were the additional features of left undescended testis and inguinal hernia, right-sided spermatic cord hydrocele and epigastric hernia which have not been specifically documented in previous reports.

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. The presence of certain distinct cutaneous features and absence of CNS and/or subcutaneous lipomas associated with alopecia, distinguish all the above mentioned disorders from ECCL. ECCL is an important syndrome for the dermatologist to recognize on encountering intracranial and cutaneous lipomas in a patient with epibulbar choristomas and CNS abnormalities. The high frequency of neurological manifestations in these patients makes radiological imaging and neurological consultation mandatory.

Histopathological examination of skin and eye lesions will further differentiate this condition from epidermal nevus syndrome, nevus sebaceous syndrome and Goltz syndrome.

There is no specific treatment for ECCL. In our patient, the management was symptomatic. Analgesics and anticonvulsants were used to control convulsions and headache while surgical correction of ocular defects and cutaneous lesions (lipomas and connective tissue nevi) was performed for cosmetic improvement. Thus, the unusual pattern of distribution, extensive CNS calcification and additional features set this patient apart from the previously reported cases of ECCL.

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