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A Rare Case of Encephalocraniocutaneous lipomatosis

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Abstract: **Encephalocraniocutaneous lipomatosis is a very rare neurocutaneous disorder. A 30 year old lady came to OPD presenting with small periocular papules, epibulbar choriostomas along with a lipomatous swelling in the frontotemporal area and a patchy alopecia on the scalp. On imaging of the brain it showed dilatation of the ventricle along with cerebral atrophy of the right side. It was finally diagnosed as a case of encephalocraniocutanous lipomatosis based o the Moog's criteria.**

Keywords: **encephalocraniocutaneous lipomatosis, neurocutaneous disorder.**

1. INTRODUCTION

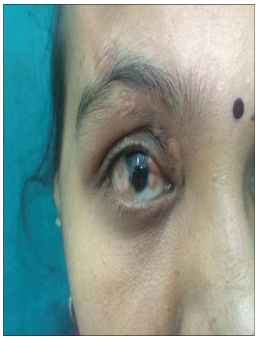
Encephalocraniocutaneous lipomatosis is a very rare neurocutaneous disorder. It was first reported by Haberland and Perou in the year 1970 and hence it is also called as the" Haberland syndrome" and "Fishman's syndrome". It is very sporadic in nature with ocular manifestations like choriostoma and subcutaneous papules, dermatological features like subcutaneous lipomas and auricular tags and neurological abnormalities like intracranial lipomas. The symptom of ECCL very closely resembles that of Goldenhar syndrome and Oculocutaneous syndrome and hence its diagnosis can be challenging.

2. CASE REPORT

A 30 year lady came to the eye OPD with complaints of a small growth seen on her right eye. It has been there since 2 years, asymptomatic and not increasing in size. On further assessment the visual acuity was found to be 6/6 in both the eyes. Examination of the anterior segment showed a diffuse yellow growth both on the nasal and temporal side of the bulbar conjunctiva more in favour of a lipodermoid. There were small papular lesions seen in the periocular area, near the canthus and along the upper lid margin. Fundus examination was done and it revealed a slightly enlarged disc with a peripapillary stippling. The left eye appeared to be normal.

A diffuse swelling was noticed in the right frontotemporal region. It was subcutaneous in nature, measuring 8*6 cm , soft in consistency with well defined margins. The skin over the swelling appeared normal. It resembled a typical lipomatous subcutaneous swelling.

A skin tag was noticed behind the right ear. More importantly the lady developed a tongue shaped patch of alopecia extending from the forehead near the right frontotemporal region.



A C T scan of the brain was done and it showed dilatation of the right lateral ventricle along with increased CSF fluid.

A complete cardiac and neurological evaluation was done and it was normal. The lady had a normal birth history, with normal developmental milestones. No history of seizures in the past

Figure 1: small papular lesions in the periocular area along with diffuse yellow growth on the nasal and bulbar conjunctiva .

Figure 2: diffuse swelling in the right fronto temporal region along witha tongue shaped patch of alopecia.

MODIFIED MOOG'S CRITERIA FOR DIAGNOSING ENCEPHALOCRANIOCUTANEOUS LIPOMATOSIS.

SYSTEM	MAJOR CRITERIA	MINOR CRITERIA
Eye	1. Choriostoma with or withour associated abnormalities	1.corneal or anterior chamber anomalies 2. ocular or eyelid coloboma 3. calcification of the globe.
Skin	1. Proven nevus psiloliparius(NP) 2. 2.Probable NP and >-one of the minor criteria2-5 3. >- two of the minor criteria 2- 5	1.possible NP 2. patchy non scarring alopecia 3.subcutaneous lipomas 4.Focal skin aplasia/hypoplasia 5.skin tags on eyelids
Central nervous system	1. Intracranial lipomas 2. Intraspinal lipomas 3. Two of the minor criteria	1.abnormal intracranial vessels 2.arachnoid cyst /abnormality of meninges 3.completet /partial atrophy of hemisphere 4.a porencephalic cyst 5.dilated ventricles 6.calcification
Others	1 .Jaw tumours 2.Coarctation of aorta 3.Multiple bone cysts	

APPLICATION OF DIAGNOSTIC CRITERIA

Definite case	1. Involvement of three systems with major criteria >2 2. Involvement of three systems,proven NP or possible NP and >1 minor skin criteria 2-5 3. Involvement of two systems with major criteria >2,one of which is proven NP or possible NP along with >1 minor skin criteria 2-5
Probable case	1. Involvement of two systems, major criteria in both 2. Two systems involved, proven or possible NP.

Based on Moog's modified criteria our patient had one major eye criteria(epibulbar choriostoma), one major skin criteria(three minor criteria- alopecia, lipomatous swelling in the fronto temporal region, auricular tags)and one major CNS criteria(two minor criteria- dilatation of the lateral ventricle, cerebral hemi atrophy).

The lady was followed up again after one year and then after 3 years .The vision still remained 6/6 in both eyes. The anterior and posterior segment findings were the same as the initial presentation. Eye pressure was measured and as found to be 15 mm Hg and 12 mm Hg in the right and left eye respectively. Dermatological findings were similar. The patient did not have any systemic abnormalities and was leading a normal life.

3. DISCUSSION

ECCL is a very rare neurocutaneous disorder with varied ocular , dermatological and neurological abnormalities. Though the exact aetiology still remains unknown, studies have suggested that a mosaic rat sarcoma gene (RAS)opathy could be considered . A group of developmental disorders caused by mutation in the RAS subfamily- mitogen activated protein kinase pathway was responsible. It has also been reported that mutation in the gene encoding for the fibroblast growth factor receptor 1 is also a possibility.

It has ocular, dermatological and neurological manifestations of which ocular abnormalities are most common. It includes epibulbar choriostomas and small periocular skin tags. Apart from these other findings include eye lid coloboma, high myopia, pallor of the optic nerve, hypertelorism, epicanthus inversus, , cicatricial ectropion of the eye lid, proptosis due to fatty infiltration of the orbit.

Skin manifestations include subcutaneous lipomas, an area of non scarring alopecia, lipomas in the neck,lumbar,axillaryregion. Periocular papules have also been noted. CNS abnormalities include intracranial lipomas, intraspinal lipomas, intracranial cysts, hydrocephalous, atrophy of the cerebral hemispheres.

There have been reports where ECCL have been diagnosed as Goldenhar syndrome until the neurological manifestations were to be seen. If this condition is diagnosed early it will help us to rule out co exiting systemic abnormalities and also start genetic counselling at the earliest.

In our case the patient was diagnosed to have ECCL based on Moogs's criteria. She was able to lead a normal life and do her day to day life activities. Hence it is important for physicians to consider and keep in mind a case of ECCL ,which is very rare though , in patients having mainly alopecia and epibulbar dermoids.

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