Hemimegalencephalic Variant of Epidermal Nevus Syndrome: Case Report

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Abstract- The epidermal nevus syndrome (ENS) is an uncommon neurocutaneous disorder in which epidermal nevi are found in association with congenital abnormalities of the brain, eye, and/or skeleton. The association of epidermal nevi and neurologic abnormalities was comprehensively described by Schimmelpenning 4 in 1957. Pavone et al. 7 (1991) identified a homogeneous variant of ENS with hemimegalencephaly, gyral malformation, mental retardation, seizures and facial hemihypertrophy.

We found 57 previously reported cases with the hemimegalencephalic variant of epidermal nevus syndrome, in which the most frequent associated features are severe epilepsy, in about half of cases with neonatal onset, mental retardation/developmental delay, ocular/visual involvement, and facial abnormalities. Here we report a 11-year-old boy with the neurologic variant of ENS with hemimegalencephaly, facial asymmetry, seizures and mental retardation.

Index Terms- Epidermal nevus syndrome, Hemimegalencephaly, Epilepsy, Mental retardation; Organoid nevus syndrome;

I. Introduction

Epidermal nevus syndrome (ENS) is an uncommon neurocutaneous disorder in which epidermal nevi (EN) are found in association with congenital anomalies of the brain, eye, and/or skeleton.[1] Most of EN are present at birth and frequently follow the lines of Blaschko. Their incidence has been reported to range from 1 to 3 per 1000 live births, without gender predominance.[2,3]. In 1957 Schimmelpenning and subsequently Feuerstein and Mims described the association between these skin lesions and central nervous system involvement, additionally reporting ocular and skeletal abnormalities. The ENS, also known as 'organoid nevus syndrome', 'linear sebaceous nevus syndrome', 'Schimmelpenning–Feuerstein–Mims syndrome', 'Solomon syndrome', and 'Jadassohn's nevus phakomatosis', is now considered a heterogeneous group of disorders [4].

Hemimegalencephaly (HME) is the commonest brain malformation associated with ENS. The association of HME and EN together with the presence of hemifaciallipoma, epilepsy and mental retardation was first described in 1955 by Gross and Uiberrak [5,6]; subsequently, Pavone and colleagues found out further 17 patients with similar neurological findings in a literature review of 63 ENS cases.[7]

II. Case Report

This 11 year old boy was the second of three children born to unrelated parents. The other two siblings were reported to be healthy. During Pregnancy, mother took tablets for aborting this child. Delivery was Institutional and uneventful. Birth weight was 2.5 kg. Developmental milestones were delayed in all domains. Child started to have right focal seizures from six months of age and is still persisting inspite of multiple antiepileptics. Child is now on adequate dose of four antiepileptics.

Child has facial asymmetry, weakness and motor clumsiness of the right arm and legs. Tone and power was reduced in corresponding limbs. Babinski sign was positive on right side.

Skin examination revealed a epidermal nevus on left forehead deviating toward left bridge of nose Fig.(C). Neurological examination revealed a macrocephaly (head cirumference = 54.5 cm, left (30cm) > right (24.5cm); Hypertrophy was observed in left upper and lower limb. Left thumb size was bigger than right thumb. BERA revealed left sided profound sensori-neural hearing loss.

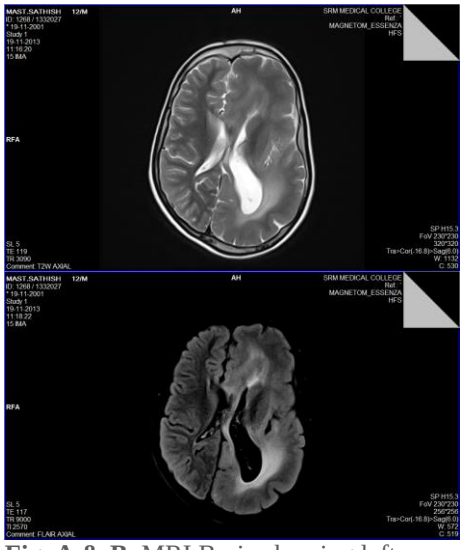

Fig. A & B. MRI Brain showing left hemimegalencephaly with broad based gyri (pachygyria), gliotic changes in left cerebral matter and colpocephaly [involving occipital horn of left lateral ventricle].

Binnet-kamet Scale demonstrated a moderate mental retardation (I.Q- 50). Ophthalmologic examination was unremarkable. Oral examination showed enamel hypoplasia with crowding of teeth. The EEG showed a severe subcortical seizure activity. Magnetic Resonance Imaging showed large left cerebral hemisphere with pachygyria and colpocephaly, altered signal intensity of white matter in left cerebral hemisphere. (Fig. A & B). Skin biopsy of the lesion showed thickening of epidermis resembling squamous papilloma.

III. Discussion and Conclusions

Epidermal nevus a congenital hamartoma of embryonal ectodermal origin is classified on the basis of component; namely sebaceous, apocrine, eccrine, follicular, or keratinocytic. An estimated one third of individuals with epidermal nevi have involvement of other organ systems; hence, this condition is considered to be an epidermal nevus syndrome (ENS).

ENS is a congenital neurocutaneous disorder characterized by linear epidermal nevus with significant involvement of the nervous, ophthalmologic, and/or skeletal systems.[2].

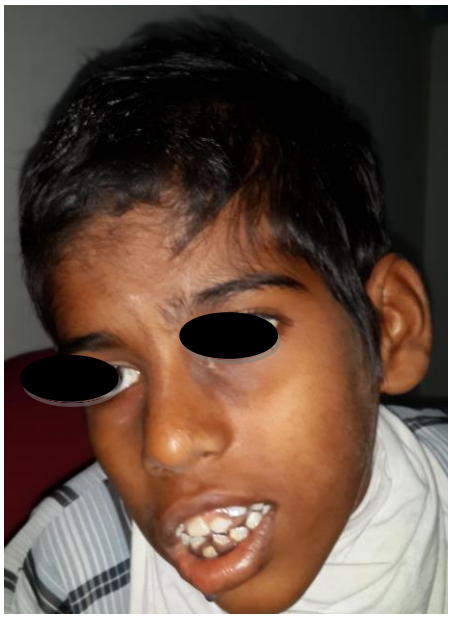

Fig.C : Showing epidermal nevus on left forehead deviating along left bridge of nose.

Clinical manifestations include Linear epidermal nevus, which may be bilateral, asymptomatic patches or plaques, with the head and the neck, as well as the trunk, being the most common sites. The lesions may have verrucous appearance and lack erythema and pruritis.

Neurological manifeststion include hemiparesis contralateral to skin and hemimeglencephaly lesion, mental retardation, seizures, and movement disorder. Eye manifestations include lipodermoid, coloboma, choristoma. Skeletal manifestations include kyphoscoliosis. Intracranial and/or intraspinal lipomas may occur. The basis of the cause may be the activation of an autosomal dominant lethal mutation that survives by mosaicism. These cells might survive only by being adjacent to normal ones.

Diagnosis: MRI can be used to evaluate intracranial involvement. MRIs may show cerebral atrophy, dilated ventricles, hemimegalencephaly (usually ipsilateral to the major skin lesions and contralateral to neurologic deficits), pachygyria, or enlarged white matter. EEG findings are abnormal in approximately 90% of patients. In almost all patients who had focal paroxysmal electroencephalographic abnormalities, the epileptiform focus was ipsilateral to the major skin lesions. Pavlidis E et al [8] in his case report and literature review has reported 57 cases of ENS in which the hemimegalencephaly was ipsilateral to the skin lesion in majority of children similar to our case report.

Treatment: No ideal medical therapy for the cutaneous lesions of epidermal nevus syndrome exists. Therapy is often challenging. Epidermal nevi are usually resistant to topical and intralesional steroids, dithranol, topical retinoids, and cryosurgery. Topical calcipotriol may be effective. The concomitant skeletal and ocular defects such as cataracts and lid anomalies can usually be surgically repaired. The CNS defects may lead to epilepsy which is usually resistant to antiepileptics. Periodic EEG and skeletal radiological analysis may be important to the long-term care of the child.

Genetic counselling

The child and/or the family should be reassured that ENS is not a genetic disorder that can be passed to future children.

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