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Case Report

Epidermal nevus syndrome associated with ocular symblepharon and gum hypertrophy - A rare variant

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Abstract

Epidermal nevi are congenital hamartomas of embryonal ectodermal origin classified on the basis of their main component. The component may be sebaceous, apocrine, eccrine, follicular or keratinocytic. An estimated 1/3 of individuals with epidermal nevi have involvement of other organ systems; hence this condition is considered to be epidermal nevus syndrome. Solomon defined epidermal nevus syndrome as a sporadic neuro-cutaneous linkage of congenital ectodermal defects in the skin, brain, eyes and skeleton, now also referred to as Solomon syndrome. The syndromes are uncommon. Mortality and morbidity are associated with systemic anomalies.

Key words

Epidermal nevi, Congenital, Hamartomas, Sebaceous, Neuro-cutaneous.

Introduction

Epidermal nevus syndrome is a disorder first proposed in 1968 by Solomon, et al. [1]. Although no clear definition has yet been presented, it is generally understood to be a rare, non-hereditary congenital disorder involving an epidermal nevus, anomalies of CNS, eyes and osseous tissues [1, 2]. The ocular complications are present in about 10-30% cases of syndrome. We describe herein a case of epidermal nevus syndrome associated with ocular symblepharon and gum hypertrophy on one side of face. Although various complications have been reported [2, 3], the present findings have not previously been described.

Case report

A 10 years old girl born from non-consanguineous parents presented with dark, verrucous hyper keratotic plaques over the left side of scalp and face and swelling of left eye since birth. Her birth weight was normal. The

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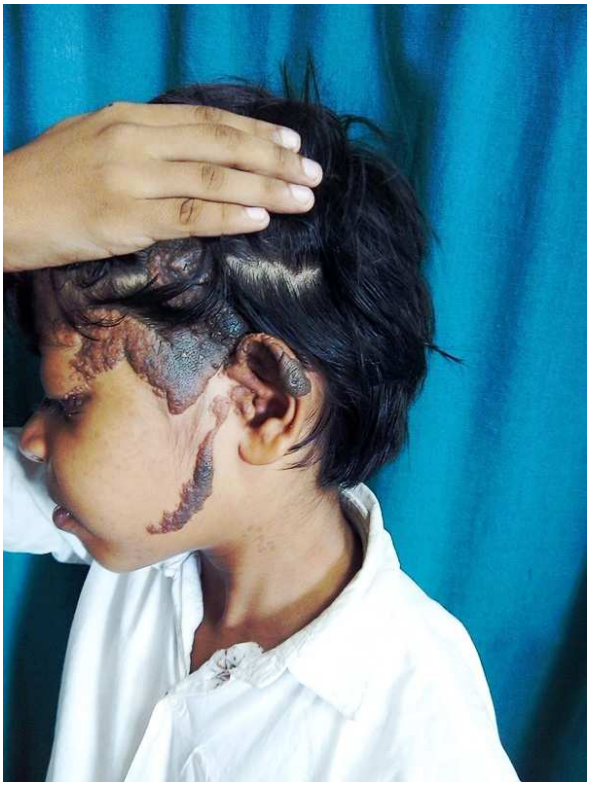
reddish mass in the left eye was increasing in size since then, causing her vision impairment. The other siblings were normal. The parents had also never suffered from any form of follicular hyperkeratosis or such ocular illness. There was no apparent family history of congenital disorder.

On cutaneous examination, there was involvement of scalp with hyper pigmented, hyperkeratotic verrucous plaques extended up to the left side of face. **(Photo - 1)** On squeezing the scalp lesions, few lesions were showing white cheesy material coming out of it.

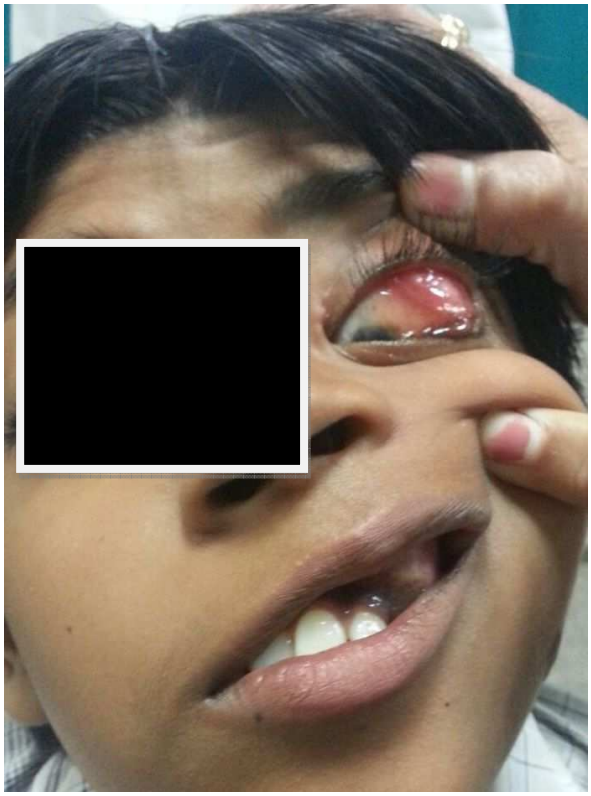
Photo – 1: Sebaceous nevus over the left side of scalp extended up to the face.



Ocular examination revealed a large reddish brown mass on the superior temporal quadrant of the conjunctiva. The mass was soft, non reducible and non compressible and adherent to palpabral and bulbar conjunctiva. Cornea revealed clouding. **(Photo - 2)** There was beginning of the similar lesions on the right eye as well. There was maxillary and gum hypertrophy over the left premolar area. **(Photo - 3)**

Photo – 2: Left eye showing symblepharon.

Photo – 3: Gum hypertrophy over the left premolar area.



CT imaging reveals eccentric expansile cystic lesion in the left half of maxillary bone involving incisor, canine and first premolar tooth. The

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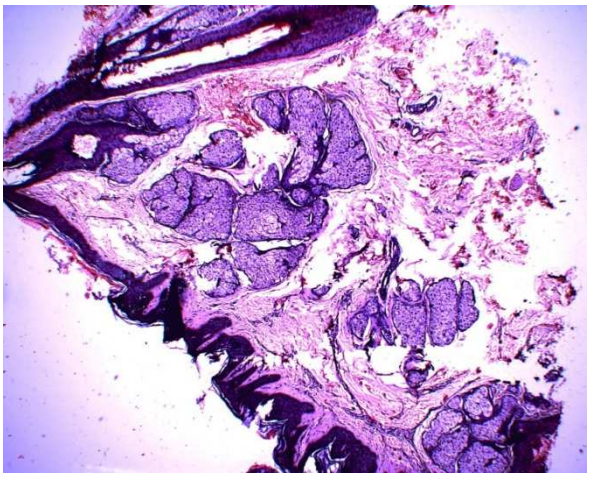
lesion showed dense internal calcification within it, likely as odentogenic keratocyst. There was unilateral proptosis of left eye ball inferio-laterally with increased supra and retro bulbar fat. Focal choroidal calcification in supero-medial left eyeball and medial aspect of right eyeball. Left preseptal thickening and soft tissue thickening in left temporal and occipital region were as likely soft tissue lesion. Bossing of left frontal bone was present.

In skin biopsy, the epidermis shows mild acanthosis and papillomatosis with immature and abnormally formed pilosebaceous units. Histological features were consistent with sebaceous nevus. **(Photo – 4, Photo - 5)** Conjunctival biopsy showed conjunctiva with severe inflammation. On general physical examination, the child was moderately built and nourished. Vitals were normal. All routine investigations were within normal limits.

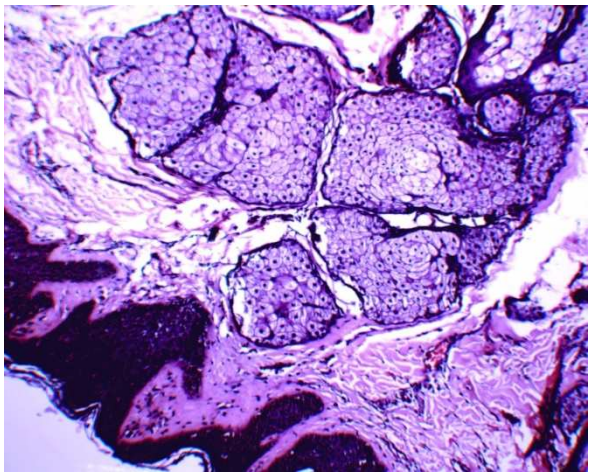
Photo – 4: The epidermis shows mild acanthosis and papillomatosis with immature and abnormally formed pilosebaceous units. (H & E, 4X)



Discussion

The association of epidermal nevi with abnormalities in other organ systems has been described in the literature for more than100 years. In 1895, Jadassohn described localized congenital lesions on the face and scalp, naming them as “organ nevi” [4]. Epidermal nevus syndrome demonstrates a variety of phenotypes. Different tissues have different embryonal origin. Skin arises from surface ectoderm; bone tissue from mesoderm and nervous tissue including eyes arise from neuro-ectoderm. Ocular complications described include tumors of eyelids, conjuctiva, cornea or sclera; coloboma of eyelid, ossification of sclera, corneal and vitreous opacities [5]. As the eye contains various tissues originating from the surface ectoderm, mesoderm and neuro-ectoderm, abnormal differentiation may take place in ocular tissue associated with epidermal nevus syndrome.

Photo – 5: Skin biopsy exhibiting lobules of abnormally formed pilosebaceous units. (H & E, 10X)



The skin lesions of this patient fulfil the criteria of an epidermal nevus because they represent a visible, circumscribed and long lasting skin disorder suggesting mosaicism. It appears difficult, however to categorize this anomaly within the group of established type of epidermal nevi. Our case closely resemble with the Schimmelpenning syndrome. Sebaceous nevus is the hallmark of the syndrome as

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present in our case. This phenotype was comprehensively described by Schimmelpenning in 1957 [6]. His patient had epileptic seizures, deformity of skull, coloboma of eyelid, symblepharon, corneal opacity and ipsilateral nevus sebaceous of scalp and face. The syndrome always occurs sporadically. This may be the best explained by the action of a lethal mutation that survives by mosaicism [7]. The phenotype is not heritable because the underlying mutation, when present in zygote, will lead to death of embryo. Cells that carry mutation can survive only in close proximity to normal cells.

Conclusion

It is a rare case of sebaceous nevus with ocular involvement and gum hypertrophy under the umbrella of epidermal nevus syndrome.

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