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Familial en Coup de Sabre With Neuropsychiatric Manifestations: A Case Report

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Abstract

Observation: En coup de sabre is a localised collagen vascular disorder which may sometimes be associated with deeper involvement of underlying neurological involvement. It is multifactorial in causation and genetic predisposition has a definite role to play. A classical case of en coup de sabre is described in a 13 year old boy with interesting neuropychiatric associations. The presence of similar lesions in his mother makes the case even more interesting. Though psychiatric associations are less commonly described with en coup de sabre, it should be looked for. The familial occurrence highlights the genetic and probably environmental causation of this disease.

Introduction

En coup de sabre or linear morphea affecting the scalp and forehead is often associated with underlying bone involvement and sometimes with neuropsychiatric manifestations. There are few reports of familial occurrence. We present a rare case of familial en coup de sabre (ECS) in mother and son, with the son having psychiatric manifestations. Neuroimaging showed falx cerebri calcification and cortical thinning.

Case Report

A 13 year old boy was brought by his mother to our clinic with complaints of a linear hyperpigmented plaque on right side of forehead since 6 years. The lesion started with itching, hyperpigmentation, binding down and mild redness and slowly extended in size for first 3 years. Three years back, the lesion became static and later the binding down started to decrease. Since last one and half year the lesion was asymptomatic and there was no increase in size. The patient had taken treatment in the form of topical steroids, tacrolimus, oral dapsone for 4 -6 months each and oral steroids for a span of one month during the initial 4 years. He was off all treatment for his skin lesion for the last 2 years. He was currently concerned about the persistent hyperpigmentation and depression over scalp.

On examination, there was involvement of right side of forehead, just lateral to the midline, in the form of a linear, dark brown, depressed slightly

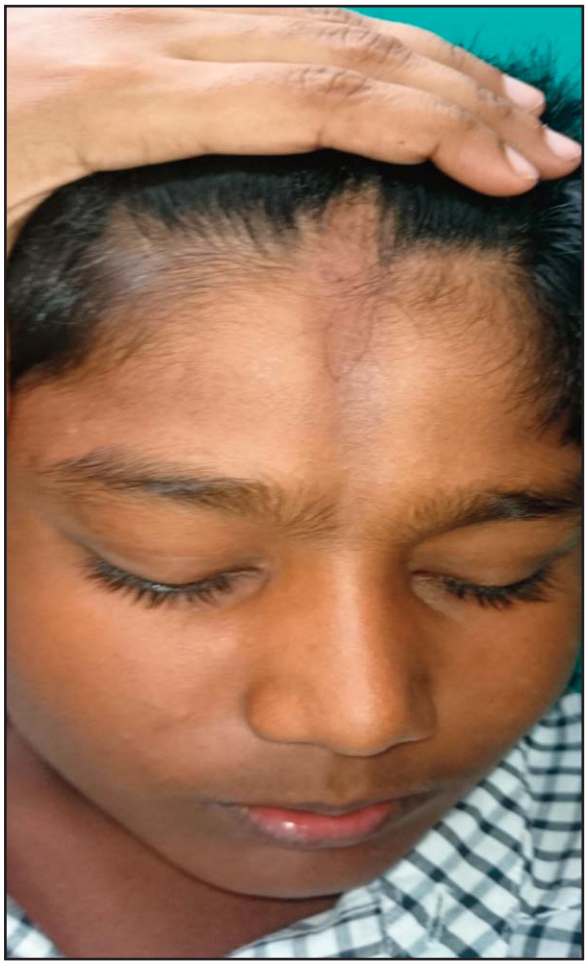

Figure 1. Hyperpigmented, depressed linear plaque on left side of forehead extending on to scalp in patient

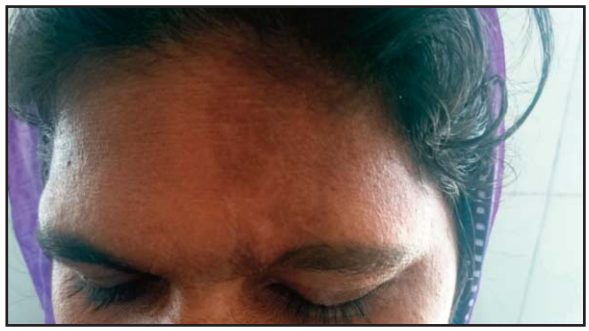

Figure 2. Hyperpigmented depressed plaque on left side of forehead in mother with a sharp central cut off

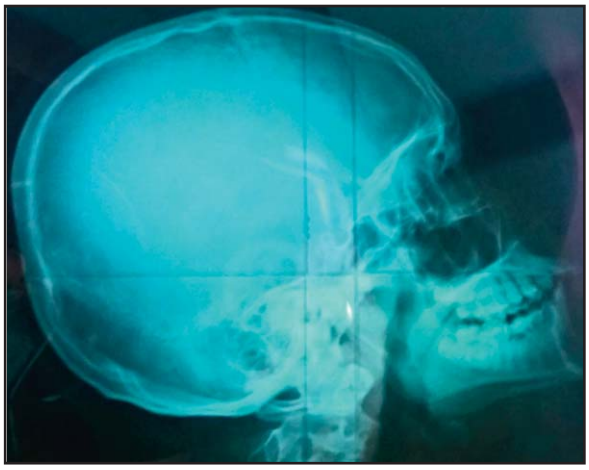

Figure 3. X ray skull [lateral view] showing depression of frontal skull and corpus falx cerebri calcification

bound down plaque measuring 6 x 0.5 cm and extending from hair line to supraciliary ridge (**Figure 1**). There was bony depression palpable underlying the plaque. There was no erythema or edema in the lesion. Facial atrophy and ocular involvement were absent.

Incidently, on reviewing the medical records of the patient, it was found that he was having violent outbursts, episodes of self harm, rage and disinhibition since last 4 years. He was constantly getting into fights at school and using abusive language at home. There was no history of seizures, recurrent headaches or any motor weakness. Neurological examination revealed no abnormalities. He was under psychiatric assessment and was diagnosed as behavioural disorder. He was started on tablet divalproex sodium 250 mg daily and along with tablet aripiprazole 5 mg daily since 3 months, with which he had around 70 – 80 % improvement in his symptoms. Intelligence quotient [IQ] testing was done which showed a normal IQ of 110. It was also noticed that his mother had hyper pigmented plaque on left side of forehead with a sharp midline cut off (**Figure 2**). On probing, she reported these lesions to have started at an age of 15, was more bound down and hyper pigmented to start with, slowly decreased in severity over last 20 years. She also had 2 other hyperpigmented bound down plaques on lower face and on neck. There was no facial atrophy or neurological complaints in the mother. Since she was not concerned about her lesions and refused further evaluation, biopsy could not be done.

Skin biopsy in the boy showed thickened sclerotic collagen bundles with mild lymphohistiocytic infiltrate, suggestive of morphea. X ray skull showed depression of frontal bone, calcification of falx cerebri and a small sella turcica (**Figure 3**). Magnetic reasonance imaging [MRI] showed no parenchymal lesions.

With these clinical features and radiological findings, a diagnosis of familial ECS with central nervous system involvement was made. Since the skin lesion was already worn out and the behavi-

Table 1. Neurological and Psychiatric Associations of En Coup de Sabre

Epilepsy – most commonly complex partial seizures
Headache including migraines and hemiplegic migraines
Focal neurological deficits
Movement disorders
Intellectual deterioration
Facial palsy
Trigemial neuralgia
Behavioural changes
Brain cavernomas

oural problems were under control, no active pharmacological intervention was done. The patient was counselled regarding the course of disease and referred to a higher centre for autologous fat transfer.

Discussion

Linear morphea affecting the scalp and forehead, otherwise known as En coup de Sabre (ECS) is a relatively uncommon type of morphea. It usually starts in first or second decade and is more common in females. It is characterised by hyperpigmented depressed bound down plaque with sharp margins. Underlying bone may be involved.

The exact etiology of ECS is known. It is considered primarily to be an autoimmune disease with doubtful roles played by viruses and bacteria like Borrelia. Fibroblast proliferation leading to increased deposition of collagen and endovascular inflammation constitute the major pathogenetic factors. Biopsy shows normal, flattened or atrophic epidermis with homogeneous, eosinophilic and dense collagen in dermis. Scanty preivascular inflammatory cell infiltrate may be seen. Elastic tissue is reduced and there is scarcity of dermal appendages and fat.

Familial ECS is relatively rare with a 4.7% twin concordance of 1.6% frequency in first degree relatives. Gene polymorphisms in various genes like BANK1, C8orfl3-BLK, IL-23R, IRF5, STAT4, TBX21, and TNFSF4, involved in immune regulation have been identified [**1**]. *Brownell* I et al described a case of two sisters with ECS in 2007 [**2**]. There are few other reports of familial occurrence of morphea in other sites [**3, 4, 5, 6**].

Various systemic associations have been described with ECS. It may vary from minor associations like malaise, arthralgia and fatigue to more severe ophthalmologic, rheumatologic and neurological involvement. Neurological involvement in ECS is rare and may sometimes precede the cutaneous manifestations. The neurological manifestations described in ECS are tabulated. (**Table 1**)[**7,8**]. Neuropsychiatric symptoms including behavioural changes and intellectual deterioration are described in 15% patients according to literature. Headache may be a symptom in upto 35% patients.

Neurological involvement in ECS is considered to be secondary to perivascular inflammation and vasculitis which leads to focal cerebral necrosis [**5**]. The most common radiological change seen is intraparenchymal calcification. Other changes include outer diploe thinning, cerebral atrophy, subcortical calcifications, meningocortical changes, T2 hyperintense images on magnetic reasonance imaging and hippocampal atrophy. Nonspecific vascular alterations, intracranial aneurysms and brain cavernomas may be identified by angiograms. Our patient showed corpus callosum calcification and thinning of outer diploe which corroborates our clinical diagnosis. The corpus callosum calcification is the first report of its kind.

Various treatment modalities have been tried in ECS with neurological involvement. Oral or parenteral steroids seem to be the most commonly used treatment in severe neurological

involvement but there are no randomised control trials in this regard [**6**]. Since our patient had an inactive skin lesion with excellent control of his psychiatric complaints, no other medication was added.

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