

Sturge Weber Syndrome with Bilateral Port-Wine Nevus

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Abstract

Sturge-Weber syndrome is a rare neurocutaneous syndrome characterized by port-wine stain, seizures and intracranial calcifications. The present case had bilateral port-wine nevus, generalised tonic-clonic seizures and right sided intracranial calcifications involving right temporo-parieto-occipital lobe regions with prominent choroid plexus.

Keywords: Sturge Weber Syndrome; Port-wine stain; Intracranial calcifications

Introduction

Sturge-Weber syndrome is a rare disorder that occurs with a frequency of 1: 50,000 [1]. It is a sporadic neurocutaneous disease characterized by facial port-wine stain, ocular abnormalities (glaucoma and choroidal haemangioma) and leptomeningeal angioma most often involving occipital and posterior parietal lobes [2]. This syndrome consists of constellation of symptoms and signs including a facial nevus, seizures, hemiparesis, intracranial calcification and mental retardation [3]. The disease's most frequent feature is the presence of epileptic crisis ranging from 74% to 90% of the cases [4].

Sturge Weber Syndrome (SWS) was first described by Schirmer in 1860 and later more specifically by Sturge in 1879, which associated dermatological and ophthalmic changes of the disease to neurologic manifestations. Weber in 1929 complemented it with the documentation of radiologic alterations seen in these patients [5].

Encephalofacial angiomatosis [6] or encephalotrigeminal angiomatosis are used as synonyms of the syndrome as angiomas involve the leptomeninges and skin of the face typically in the ophthalmic and maxillary distributions of the trigeminal nerve. Developmental disorders are more common when angiomas are bilateral [7].

Case Presentation

A one and a half year old boy was brought to Department of Paediatrics with complains of left sided tonic clonic seizures and facial twitching associated with frothing from mouth and altered sensorium lasting for 10-15 minutes which occurred 2 days back and controlled with oral phenytoin. He had no similar episodes of seizures in the past. The child was born full term normal delivery to non consanguineous marriage. There was the presence of well defined red to purple nevus over bilateral face. There was no family history of similar disease.

Physical examination revealed a febrile child with pulse rate of 126/minute, respiratory rate of 32/minute and his blood pressure was 76/50 mmHg. There was port-wine stain involving bilateral face involving ophthalmic, maxillary and mandibular division of trigeminal nerve (Figure 1). He was active and taking feeds well. Neurological examination showed no focal deficits. Ophthalmic examination revealed no abnormality. Total and differential counts, blood sugar, sodium, potassium, calcium, phosphate were within normal range. Contrast Enhanced Computed Tomography (CECT) head showed multiple foci of calcifications (linear, curvilinear and punctuate) with abnormal meningeal and gyral enhancement in right temporo-parieto-occipital lobe region with slightly enlarged ipsilateral choroid plexus (Figure 2).



Figure 1: Clinical photograph of the face showing bilateral port-wine nevus.



Figure 2: Cranial CECT showing right side intracranial calcifications.

So based on the presence of seizure, port wine stain over bilateral and calcification, the diagnosis of Sturge Weber syndrome (SWS) was considered.

Discussion

Sturge-Weber syndrome is a rare neurocutaneous disorder and is

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referred to as complete when both central nervous system and facial angiomas are present and incomplete when only one area is affected without the other [7]. Our patient had complete Sturge-Weber syndrome.

The inheritance of Sturge-Weber syndrome is sporadic [6] and it occurs with a frequency of 1: 50,000 [1]. Both the genders are equally affected and no racial differences have been reported [8]. Although the precise pathogenesis is unknown, it is thought to be the result of anomalous development of the primordial vascular bed during early stages of cerebral vascularisation [3].

Clinical manifestations are differentiated into three categories:

Cutaneous features

The facial port wine nevus, the most common cutaneous manifestation, is usually present at birth and localised along the trigeminal areas of the face, but in some cases it can be seen over trunk, oral mucosa or pharynx. Unilateral involvement accounts for 71.6%, bilateral nevus for 28.4%, absent nevus for 5% [2]. Our patient has bilateral nevus as seen in (Figure 1). Children with a bilateral nevus may have a greater chance of bilateral brain involvement and thus more likelihood of neurological impairment and earlier onset of seizures [9,10].

Neurological involvement

Most patients have normal neurological functions for several months or even years after birth. Seizures, the most common neurological disturbance, occur in 72% of patients with unilateral and 93% of those with bihemispheric involvement [9]. Focal motor seizure or generalised tonic clonic seizure predominates initially. Patients with refractory epilepsy are much more likely to be mentally retarded than those with milder seizures [9].

Ophthalmologic complications

Glaucoma and buphthalmos of the ipsilateral eye are the common complications. Glaucoma develops in up to 42% of patients whose facial cutaneous angioma is adjacent to eye [11]. Glaucoma occurs independently of intracranial involvement. Our patient did not have Glaucoma. However, screening for glaucoma is probably justified because early diagnosis and aggressive management of glaucoma are imperative [12].

Neuro-imaging helps to distinguish children with SWS from those with isolated facial lesion. MRI with gadolinium enhancement is at present the best technique [13,14]. And more efficient in the detection of the radiological findings related to the clinical status than CT scan. The skull radiograph shows occipito-parietal region in most patients. This characteristically assumes a serpentine or "railroad-tract" appearance.

The management is multifaceted. The patients with well controlled seizures and normal or near - normal development, the management is conservative. Seizures in our case were controlled with oral phenytoin sodium (5mg/kg/day). However, there is increasing evidence that hemispherectomy or lobectomy may prevent the development of mental retardation in patients with recalcitrant seizures. Flash-lamp-pulsed laser therapy holds considerable promise for clearing of the port-wine stain. Glaucoma is usually controlled with conservative management but if the intra-ocular pressure is persistent, combined trabeculotomy-trabeculectomy is a promising treatment for patients [15]. Special educational facilities are frequently required for developmental disabilities.

Conclusion

Our case had a bilateral nevus hence has a greater chance of bilateral brain involvement and thus more likelihood of recurrent seizures and neurological impairment. Therefore, these patients require long term follow up for evaluation of mental retardation or severe learning disability which occurs in about half of the children in later life.

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