

Case Blog

Title: Sturge Weber Syndrome Type 1 – A Case Blog with Review of Literature

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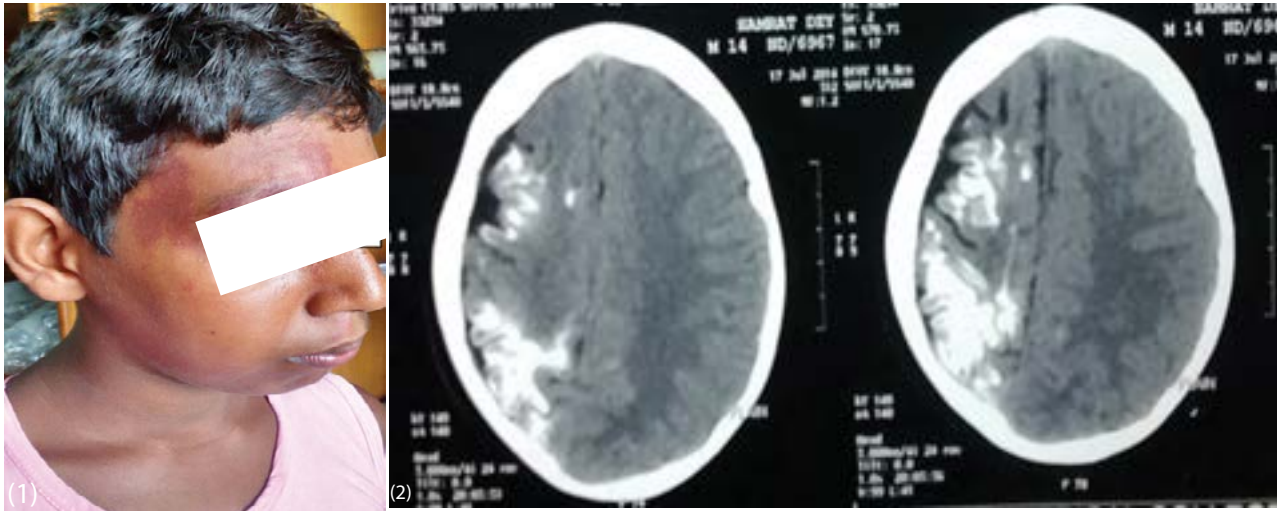


Figure 1: Showing port wine stain.

Figure 2: CT scan showing Rt. Cortical atrophy and calcifications.

Introduction

Sturge-Weber syndrome (SWS), it is named for William Allen Sturge and Frederick Parkes Weber, It is a rare syndrome, with an incidence estimated at 1 case in 20,000-50,000 persons, sometimes referred to as encephalotrigeminal angiomas [1], is a rare congenital neuro cutaneous disorders. It is one of the phakomatoses and is often associated with port-wine stains of the face, glaucoma, seizures, mental retardation, and ipsilateral leptomeningeal angioma (cerebral malformations and tumours). It is characterized by abnormal blood vessels on the brain surface. Normally, only one side of the brain is affected. This is a case report of a patient who classically presented in our Medical College with complaints of convulsion and later it was diagnosed to be Sturge Weber syndrome type 1.

Case Blog

Fourteen years old male patient presented to the emergency department with complaints of sudden onset of seizure. On examination his developmental mile stones were delayed. He also had convulsion in the left side of the body. Right side was near normal. On careful examination the boy had a reddish patch over right side of his forehead (Figure 1). The attenders were telling the mark was present since birth. The mark was totally unilateral along the branches of ophthalmic division of right trigeminal nerve. The lesion over skin was not crossing midline. it was a classical port wine stain. On examination child was drowsy, disoriented. Then immediately CT scan was planned for the patient. There were lots of malformations found in the CT scan. Haemangioma were present in the same side of the brain. These haemangioma had undergone calcification. There were multiple calcifications seen over left side CT brain. The underlying brain region was totally atrophied (Figure 2).

This was a classical presentation. The boy suddenly had an episode of epilepsy, following which he visited our emergency department. Apart from the routine CNS examination, the boy was found to have typical port wine stain. Also on the same side in

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CT scan the patient had cortical atrophy and multiple calcifications. In cases of Sturge Weber syndrome, the patients will usually present in their early childhood days with complaints of seizure. But this patient had a very late stage of presentation.

Discussion

Sturge-Weber Syndrome (encephalo trigeminal angiomatosis) is a congenital, non-familial disorder of unknown incidence and cause. It is characterized by a congenital facial birthmark and neurological abnormalities. Other symptoms associated with Sturge-Weber can include eye and internal organ irregularities. Each case of Sturge-Weber Syndrome is unique and exhibits the characterizing findings to varying degrees. The various classical findings in a case of Sturge Weber Syndrome are as follows,

(1) Facial birthmark – Port wine stain

The most apparent indication of Sturge-Weber Syndrome is a facial birthmark or “Port Wine Stain”[2] present at birth and typically involving at least one upper eyelid and the forehead. Much variation in the size of the stain has been reported and may be limited to one side of the face or may involve both sides. The birthmark, varying from light pink to deep purple, is due to an overabundance of capillaries just beneath the surface of the involved skin. In persons with dark pigmentation, country like India, this may not be taken care by the parent as a classical feature. In our case the classical port wine stain was present along the ophthalmic division of the trigeminal nerve supply oriented region. It was in the right side and also it was not crossing the midline at any level, though it is said that this port wine stain is always unilateral, cases with bilateral port wine stain is also reported in literature. Thus at the time of delivery these port wine stains have to be kept close in mind, and patients with such a presentation has to be followed up regularly.

(2) Neurological manifestations

Neurological concerns relate to the development of excessive blood vessel growth on the surface of the brain (angiomas). These are located typically on the back (occipital) region of the brain on the same side as the port wine birthmark. Seizures are one more important manifestation of this syndrome. Sometimes seizures may be intractable. Focal deficits like hemiparesis, hemianopsia may be associated with sturge weber syndrome. Some patients also have headache. In our case also major presenting complaint of the patient was headache. Mental abnormalities are more common when angiomas are presenting bilaterally. Progressive calcifications in cerebral cortex with cortical atrophy frequently develop and progress with age. In our patient also CT scan revealed cortical atrophy with calcification.

The various other associated features of these syndromes are increased pressure within the eye (glaucoma). The glaucoma is usually restricted to the eye which has the stain involvement. Enlarging of the eye (buphthalmos) can also occur in the eye which has been affected by the stain. Multiple other body organs are rarely affected in Sturge-Weber syndrome. Infants affected with Sturge-Weber Syndrome are often monitored by a paediatrician, neurologist, ophthalmologist and dermatologist. But on our patient these manifestations were not noticed. In 1992, E. Steve Roach, MD classified the SWS spectrum, delineating for the first time the varying degrees of involvement previously noted in this condition [3,4].

Type 1

The most common, this type involves both facial and leptomeningeal (brain) angioma (vascular malformations) and may involve glaucoma. Seizures usually occur within the first year of life, as a result of the brain involvement. The ocular involvement is normally noted within the first year of life. In our case though it was a late presentation, it can be classified as type 1 SWS

Type 2

This type involves a facial angioma and the possibility of glaucoma, but no evidence of intracranial disease. There is no specific time-frame for the exhibition of symptoms beyond the initial recognition of the facial PWS. Throughout the life of the individual, interrelated symptoms may manifest in glaucoma, cerebral blood flow abnormalities, headaches, and various other complications.

Type 3

This type of SWS is commonly noted to have a leptomeningeal angioma, with no facial involvement and usually no development of glaucoma.

Treatment

Treatment for Sturge-Weber syndrome is symptomatic. Laser treatment may be used to lighten or remove the birthmark. Anticonvulsant medications may be used to control seizures. When one side of the brain is affected and anticonvulsants prove ineffective, the standard treatment is neurosurgery to remove or disconnect the affected part of the brain (hemispherectomy) [5]. Physical therapy should be considered for infants and children with muscle weakness. Educational therapy is often prescribed for those with mental retardation or developmental delays, but there is no complete treatment for the delays. Thus it is a multi-modality management is necessary for sturge weber syndrome. When there is recurrent attacks of seizure neurosurgery can be considered. Sturge-Weber Syndrome is often monitored by a paediatrician, neurologist, ophthalmologist and dermatologist.

The differential is a combination of that for multiple intracranial calcifications, cerebral hemiatrophy and leptomeningeal enhancement, and therefore includes [6]:

1. Cerebral arteriovenous malformation (AVM)
2. Infection
TORCH infection
Neurocysticercosis
3. Cutaneous haemangioma–vascular complex syndrome 8
4. Healed cortical infarct
5. Radiotherapy

Conclusion

Thus when a paediatric patient is presenting to the emergency department with sudden onset of convulsion, apart from the basic CNS examination, it is always very important to look for cutaneous lesion, also proper history regarding abnormal birth mark has to be taken. If a child present with classical port wine stain and typical neurological finding diagnosis of SWS can be easily established.

References

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