Case Report

Sturge-Weber syndrome with anomalous venous drainage – An unusual manifestation of neurocutaneous syndrome

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ABSTRACT

Sturge-Weber syndrome is a rare congenital vascular disorder manifesting with a constellation of signs and symptoms including a facial capillary malformation (port-wine stain), abnormal blood vessels of the brain (leptomeningeal angioma), and associated with abnormal blood vessels of the eye leading to glaucoma. Here, we report the case of an 18-month-old child who had skin lesions on both sides of the face since birth, macrocephaly, left-sided focal seizures, and developmental delay. The child was initiated on carbamazepine at 10 mg/kg/day at admission and the dose was increased to 15 mg/kg/day. On subsequent follow-up, the child is seizure free till now.

Key words: Developmental delay, Focal seizure, Port-wine stain

Sturge-Weber syndrome (SWS) or encephalotrigeminal angiomatosis is a congenital non-hereditary condition with unknown etiology which is characterized by vascular lesions involving areas innervated by the trigeminal nerve, ipsilateral leptomeninges, and brain [1]. The disease manifests as cutaneous port-wine stain, ocular abnormalities such as glaucoma and choroidal hemangioma, and neurological abnormalities such as seizures, focal neurologic deficits, or intellectual disability. It belongs to a group of disorder known as the phakomatoses ("mother spot" diseases). The incidence of SWS is 1 in 20,000–50,000 live births [2] with no racial and gender predilection [3]. SWS is caused by the persistence of vascular plexus around the cephalic portion of the neural tube.

Cutaneous angiomas, also called port-wine stains, present ipsilaterally on the forehead and upper eyelids primarily in the distribution of ophthalmic and maxillary divisions of the trigeminal nerve. It may be bilateral or unilateral and may extend to the neck, limbs, and other parts of the body. Vascular abnormalities in SWS are seen in 40% of cases [4] and it occurs due to the lack of superficial cortical draining veins resulting in the formation of collateral pathways manifesting as dilated angioma in subependymal, transparenchymal, and deep veins [5]. Here, we report the case of an 18-month-old child with characteristic manifestations of SWS with an unusual manifestation of anomalous venous drainage.

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CASE REPORT

An 18-month-old male born out of non-consanguineous marriage to a 23-year-old primigravida with uneventful antenatal and birth history (APGAR scores at 1 and 5 min were 7 and 8) presented with complaints of the left-sided focal seizures for the past 1 month. Developmental milestones were delayed with a development quotient (DQ) of 47%. The child had no previous episodes of seizures and no family history of epilepsy.

The general examination showed that the child had occipitofrontal circumference of 52.8 cm (Fig. 1a), buphthalmos, prominent scalp veins over the forehead, and reddish discoloration of the skin (port-wine stains) involving the right side of the face, forehead, both upper eyelids, and right arm (Fig. 1b). Ocular examination was done and indirect ophthalmoscopy revealed bilateral partial optic atrophy and raised intraocular pressure (IOP) as assessed by digital tonometry. The rest of the systemic examination was unremarkable.

With a clinical possibility of SWS, a contrast-enhanced computed tomography (CECT) brain was done and showed prominent medullary and subependymal veins on the right side and enlarged ipsilateral choroid plexus along with dilation of the left ophthalmic vein (Fig. 2a and b). There was evidence of prominent subarachnoid space (Fig. 2c and d).

The child was initiated on carbamazepine at 10 mg/kg per day at admission and the dose was increased to 15 mg/kg/day. He remained seizure free during the hospital stay. For glaucoma, the timolol eye drops 0.25% were given. The child is on regular

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follow-up every 4 weeks and is seizure free till now. The IOP has reduced after medical measures.

DISCUSSION

The term "SWS" was coined by William Allen Sturge and Frederick Parkes Weber. It was first noticed by Schirmer in 1860. Sturge described various cutaneous, ocular, and neural presentations of the disease in 1869 and Weber documented various radiological changes observed in these patients in 1929 [6,7].

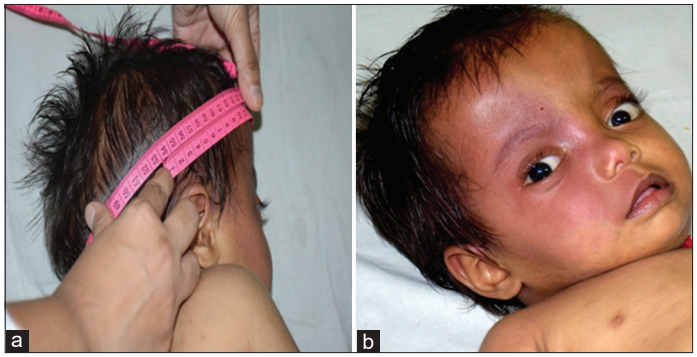

Figure 1: (a) Head enlarged with head size of 52 cm; (b) port-wine stain over both sides of the face with abnormal dilation of scalp veins

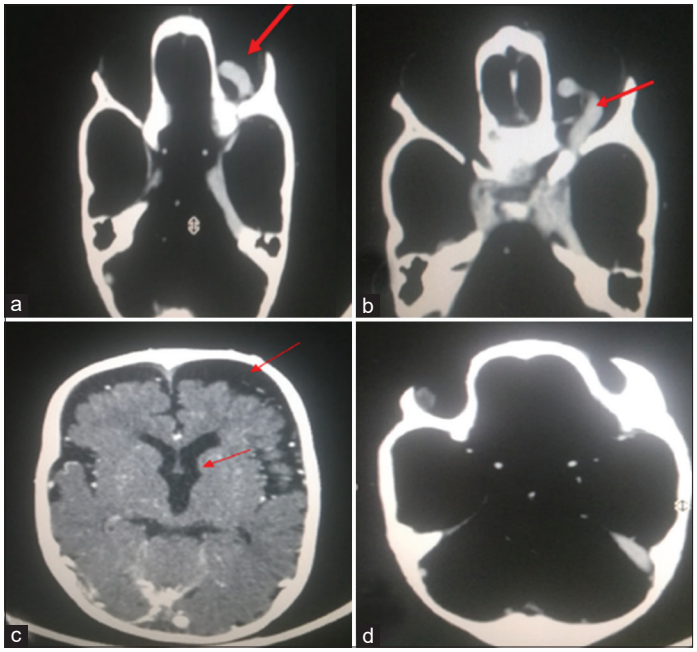

Figure 2: (a) Dilated and prominent left ophthalmic vein; (b) dilated left cavernous sinus; (c) contrast-enhanced computed tomography (CECT) head of the child showing prominent ventricular system and subarachnoid space; (d) CECT head of the child showing prominent ventricular system and subarachnoid space

Table 1: Roach scale for Sturge-Weber syndrome [8]

Type I	Both facial and leptomeningeal angiomas	Glaucoma±
Type II	Only facial angiomas	Glaucoma±
Type III	Isolated leptomeningeal angiomas	No glaucoma-

At present, the Roach scale is used to differentiate various types of SWS (Table 1) [8]. According to the Roach scale, our case is a Type I SWS. The overall incidence of SWS in those with a port-wine stain is 8–33% [5]. Buphthalmos and glaucoma of the ipsilateral eye as seen in the index case are common complications. When both central nervous system and facial angiomas are present, SWS is referred to as complete and when only one area is affected without the other, it is called incomplete SWS.

The incidence of epilepsy in patients with SWS is 75–90%. Seizures usually start in early childhood, and in most patients, seizures develop in the 1st year of life. They are typically focal and contralateral to the side of facial capillary malformation [8]. Our index case also had focal seizures contralateral to the side of facial capillary malformation. Differential diagnosis of SWS includes Rendu-Osler-Weber syndrome [9,10], Maffucci's syndrome, angina osteodystrophy syndrome, Von Hippel-Lindau disease, Klippel-Trenaunay-Weber syndrome, Parkes-Weber syndrome, and Proteus syndrome.

The involvement of the area supplied by the first division of the trigeminal nerve is pathognomonic of the disease. Ocular features of SWS include glaucoma, choroidal hemangioma, buphthalmos, or hemianopsia. According to Inan and Marcus, port-wine nevi are especially localized to the right side of the face and are detected in 87–90% of cases [11]. In about 50% of the patients, an extension of the lesion over the midline is seen and bilateral involvement can be detected in about 33% of cases [11]. In this case, the child showed nevus flammeus on both sides of the face without extension over the midline. Intellectual disability and learning disabilities are seen in later childhood in about 50% of patients [2]. In our case, the child had a global developmental delay with a DQ of 47%. Vascular abnormalities are reported in 40% of children with SWS and these vary with age. Most of the cases have reported abnormal venous drainage by 5–6 years of age [4]. Intraorally, angiomatosis involves lips, gingiva, buccal mucosa, palate, and mainly, the floor of the mouth. Oral manifestations of SWS are port-wine stain lesions of the oral mucosa and hypervascular changes [1].

Imaging of the brain (computed tomography [CT]/magnetic resonance imaging [MRI]) must be done as early as possible to look for anomalies in the venous drainage. MRI brain findings usually show the formation of angiomas in the deep venous system, choroid plexus enlargement, and abnormal deep vein enlargement [4]. Although MRI brain is the investigation of choice in SWS, in our setting due to the non-availability of MRI, CECT was done which showed prominent medullary and subependymal veins on the right side with enlarged ipsilateral choroid plexus. The left ophthalmic vein was dilated with dilated cavernous sinus and anomalous drainage of the left deep venous system into the left cavernous sinus instead of a jugular vein with the prominent ventricular system along with the evidence of prominent subarachnoid space. Dilation of the ophthalmic vein has not been reported to occur so early in age [12].

CONCLUSION

As glaucoma and seizure occur in about half of the patients, early diagnosis is critical for improvement in long-term outcome and quality of life. Seizures can be prevented by antiepileptic medications. Every child should undergo complete eye examination so that blindness and developmental delay due to visual complications can be prevented. Imaging of the brain (MRI/CT) must be done early to look for anomalous venous drainage and in case of associated hydrocephalus/hemorrhage; early neurosurgical management must be done. MRI is the modality of choice for diagnosis but in its absence, CT of the brain can be done, especially in resource-limited settings, as it is easily available in secondary and tertiary care centers these days.

REFERENCES

1. Khambete N, Risbud M, Kshar A. Sturge-Weber syndrome: A case report. Int J Dent Clin 2011;3:79-81.
2. Di Rocco C, Tamburrini G. Sturge-Weber syndrome. Childs Nerv Syst 2006;22:909-21.
3. Welty LD. Sturge-Weber syndrome: A case study. Neonatal Netw 2006;25:89-98.
4. Martí-Bonmatí L, Menor F, Mulas F. The Sturge-Weber syndrome: Correlation between the clinical status and radiological CT and MRI findings. Childs Nerv Syst 1993;9:107-9.
5. Sperner J, Schmauser I, Bittner R, Henkes H, Bassir C, Sprung C, *et al*. MR-imaging findings in children with Sturge-Weber syndrome. Neuropediatrics 1990;21:146-52.
6. Kowalska-Brocka J, Brocki M, Uczniak S, Uczniak K, Kaszuba A, Jurowski P. Sturge-Weber syndrome Type II treated with PDL 595 nm laser. Postepy Dermatol Alergol 2015;32:63-6.
7. Patil K, Guledgud MV, Sahni A. Sturge Weber syndrome-a case report. Int J Dent Sci Res 2015;3:45-7.
8. Roach ES. Neurocutaneous syndromes. Pediatr Clin North Am 1992;39:591-620.
9. Kossoff EH, Buck C, Freeman JM. Outcomes of 32 hemispherectomies for Sturge-Weber syndrome worldwide. Neurology 2002;59:1735-8.
10. Suprabha BS, Baliga M. Total oral rehabilitation in a patient with portwine stains. J Indian Soc Pedod Prev Dent 2005;23:99-102.
11. Inan C, Marcus J. Sturge-Weber syndrome: Report of an unusual cutaneous distribution. Brain Dev 1999;21:68-70.
12. Nakata Y, Yagishita A, Tsuchiya K. Imaging of Sturge-Weber syndrome: Cranial CT and MR findings. Nihon Igaku Hoshasen Gakkai Zasshi 2004;64:210-5.

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