**Sturge-Weber Syndrome With Contralateral Brain Angioma: A Unique Case Report**

**Abstract:** A 16-year-old boy with a history of epilepsy was brought to the emergency department for status epilepticus. Initially, the patient's airway, breathing, and circulation were secured and he received intravenous medications. On examination, he had left-sided facial nevus and decreased power in both his left upper and lower limbs. All other causes of seizures were ruled out and an MRI brain of the patient was ordered that revealed right-sided leptomeningeal angiomas.

**Key clinical message:**  Most facial vascular nevi in SWS are associated with an ipsilateral leptomeningeal angioma, however, contralateral angiomas can also occur in SWS. Variants of this classical presentation have been described in the literature.

**Keywords**: Sturge-Weber Syndrome; Facial Nevus; Seizures; Angiomas

**Introduction**: Sturge-Weber syndrome (SWS), also known as encephalotrigeminal angiomatosis, is a rare non-inherited congenital anomaly. It is most recognizable as a neurocutaneous syndrome associated with distinctive facial nevus flammeus (port-wine stain), leptomeningeal angioma, and episcleral angiomas leading to glaucoma. Epidemiologically, SWS is estimated at 1 per 20,000-50,000 live births.1 The pathogenesis of SWS involves a somatic mutation in a nucleotide transition in the GNAQ gene on chromosome 9q21. The exact mechanism of how this activation results in macules with violet staining and in the establishment of SWS is not yet clear.2

Classically, these patients present with congenital malformation of cephalic capillaries (i.e., birthmark on the face), mainly focal type seizures, hemiparesis or hemiplegia, intellectual disability and abnormal vasculature in the eye, leading to various ocular impairments, particularly glaucoma. Of note is the clinical significance of the nevus flammeus; when a newborn presents with a port-wine birthmark. Port wine stains are reported in about 87–90% of cases and generally occur on the right side; there is a 15-20% risk that the newborn will develop Sturge-Weber syndrome with encephalic and/or ocular involvement.3 The prevalence of seizures, however, is a more robust correlation amongst this patient cohort; approximately 75% of patients with unilateral cerebral involvement will present with seizures, and 95% of patients with bilateral involvement.4 Making a diagnosis requires clinical evaluation, and it is confirmed by imaging modalities such as plain X-ray, Computed Tomography (CT) and Magnetic Resonance Imaging (MRI) scan. Treatment is mainly symptomatic and supportive with control of seizures using anti-epileptic drugs. Surgical resection is reserved for intractable cases of seizures.5 Classically leptomeningeal angiomas in SWS occur ipsilateral to the facial nevi, this case highlights the atypical presentation of SWS in which the patient had leptomeningeal angiomas contralateral to the facial nevus.

**Case report:** A 16-year-old boy with a history of epilepsy was brought to the emergency department of a tertiary care hospital for status epilepticus. The patient's seizures started suddenly at home while he was watching television. While evaluated in the emergency room the patient had generalized tonic-clonic seizures associated with tongue biting and loss of consciousness. Further inquiry from the parents of the patient revealed that he was compliant with antiseizure medications. After initial resuscitation in the emergency room, the patient received an injection of benzodiazepine and divalproex sodium but did not respond to the medications. Blood was drawn for laboratory investigations and the patient was shifted to the intensive care unit for further management. The patient's father disclosed his son's developed seizures at 6 months of age and that progressively worsened over time in terms of frequency, intensity, and non-responsiveness to the medications. He further added that the patient had delayed speech while other developmental milestones were achieved on time. His past history is significant for glaucoma for which he underwent bilateral trabeculectomy at the age of 5 years. Family history was not significant for similar conditions. Examination showed multiple, well-defined, red to purple patches involving the face along the distribution of trigeminal nerves bilaterally, extending into the scalp as well. The left half of the body was predominantly affected. These stains were present at birth. The patient exhibited marked facial deformity secondary to soft tissue and bone hypertrophy, primarily in the maxillary region. Intraoral examination revealed gingival hypertrophy, dental malocclusion, and decreased number of teeth in the upper jaw as shown in Figure 1. No focal neurological deficit was noted. The rest of the examination was unremarkable. All the metabolic, paraneoplastic, and infectious causes of the patient's seizures were ruled out with normal laboratory tests, and a computed tomographic (CT) scan of the brain was requested for central nervous system pathology. CT scan of the brain showed dense gyriform calcifications characteristic of tram track sign, affecting the right occipital and temporal lobes as shown in Figure.2. Magnetic resonance imaging (MRI) of the brain with gadolinium enhancement demonstrated prominent leptomeningeal enhancement affecting the right temporal and occipital lobes along with ipsilateral choroid plexus enlargement. Surprisingly, the left cerebral hemisphere was unremarkable as shown in Figure. 3. Based on these physical examinations and radiologic findings i.e port-wine nevi on the left side of the face along the distribution of the trigeminal nerve, generalized seizures, and dense gyri form calcification of the right side on imaging patient was diagnosed with Sturge- Weber syndrome with atypical characteristics.

The patient received intravenous anti-epileptic medication (injection. Divalproex sodium 500 mg twice daily, injection. Diazepam 10mg, and injection. Levitiracetam 500 mg BD) to control his seizures. Parents were counseled regarding the potential triggers for seizures and the importance of annual ophthalmic examinations. Neurosurgical consultation was scheduled to consider surgical treatment of the patient’s refractory seizures. In order to address this patient's ocular and dermatological manifestations, he was referred to an ophthalmologist and a dermatologist. Subsequent evaluations were scheduled; additionally, he was prescribed oral medications, including low-dose aspirin and dose-adjusted antiepileptics upon discharge.

**Discussion:** Sturge-Weber syndrome (SWS) consists of a group of disorders that may present with neurological, cutaneous, ocular, and oral manifestations. The clinical course of SWS is variable, but it is usually characterized by progressive neurological problems such as seizures, hemiparesis, headaches, visual field deficits, and cognitive impairments as seen in the above case illustration. Other features include early handedness, gaze preferences as well as stroke-like episodes. The non-neurological symptoms of SWS include behavioral and emotional derangement, endocrine problems, and learning difficulties, cutaneous and oral manifestations.5

Cutaneous manifestation is a common presentation in SWS, notably facial nevus, usually in the distribution of ophthalmic and maxillary divisions of the trigeminal nerve. It is present at birth, remaining static throughout life. This is in contrast to the infantile hemangioma which is not present at birth. However, it is more common than port-wine stain. Facial port wine stain is associated with a 10% to 35% risk of SWS. However, they are not pathognomonic of SWS.6

Apart from the brain and skin, SWS also commonly affects the eyes with glaucoma being the commonest ocular manifestation. When present, it is almost always ipsilateral to the facial port-wine stain. 7 Other features include oral manifestations such as changes in mucosa coloration, that is, the appearance of purple-reddish macules; gingival lesions that may include a significant vascular component; macroglossia; and hemihypertrophy of palatal and buccal mucosa. Moderate-to-severe variable gingival growth may be present, which causes greater discomfort, impaired nutrition, and difficulty in hygiene, and consequently favors the accumulation of a biofilm, increasing the individual’s susceptibility to oral infections and compromising systemic health. This gingival growth may be induced by anticonvulsant medications used in treating these individuals, which usually report epileptic seizures. 8

Diagnosis of Sturge-Weber syndrome is based on the presence of clinical manifestation including neurological symptoms and cutaneous appearance, supported by imaging findings on brain MRI and or CT scan.6 A similar pattern of arriving at a diagnosis in the above index case i.e. gyriform calcification, is a common feature that is appreciated on the skull radiographs and classically described as a “tram-track sign. However, disorders known to produce cerebral calcifications such as celiac disease, encephalitis, purulent meningitis, ossifying meningoencephalopathy, and leukemia should be excluded in such cases.

SWS is classified using the Roach scale [5] into, Type 1 (Presence of both facial and leptomeningeal angiomas) Type 2 (Presence of facial angiomas alone) Type 3 (Isolated leptomeningeal angiomas; usually no glaucoma). According to the criteria, our patient fulfilled type 1. As the patient had a port wine stain with epileptic convulsions and calcification of the occipito-parietal region. Other variants of this classical presentation have been described in the literature. In another study, Siri L et al report five novel patients showing variable clinical appearance. These patients presented with SWS with leptomeningeal angioma only without facial nevus. Yallaprgada AL et al presented a case of a patient with a head and neck port-wine nevus, glaucoma, and abnormalities of the intracranial deep veins, the patient lacks any radiologic or clinical evidence of cerebral leptomeningeal angiomatosis. 9

Similar to our case, Inan et al, presented a case with unusual cutaneous manifestations. The right face and arm of the patient were smaller than the left, associated with hypotonia and moderate weakness in the right forearm. Computerized tomography of the head showed intracranial calcification in the left frontoparietal region, and a brain MRI, with gadolinium, revealed an extensive leptomeningeal angioma over the entire left hemisphere.10 In another study Widdess Walsh and co-worker describe one patient with Sturge-Weber syndrome who presented with a left-sided facial nevus, left eye glaucoma, episodes of left-sided weakness, and right-sided leptomeningeal angiomatosis by gadolinium-enhanced brain MRI thus, confirming variant of a leptomeningeal angioma contralateral to the facial nevus.11

Treatment is generally supportive and primarily aims to minimize seizure activity with anti-epileptic medications. Surgery may be offered to patients whose seizures are refractory to available medical therapies. Therefore, the mainstay of treatment is the control of seizures with the use of anticonvulsants. Early treatment of seizures is important to prevent the progression of neurological damage. Anti-epileptic drugs should be initiated early and continued for life. Low-dose aspirin has been shown to reduce the frequency of cerebrovascular events and seizures in these patients. 12 It is reported that the use of sodium valproate is the first treatment for individuals with generalized tonic-clonic seizures and shows that lamotrigine and levetiracetam would be suitable alternative first treatments. LEV combined with SV is effective in the treatment of children with epilepsy and does not increase the clinical ADR. Seizures remain difficult to control through the use of oral medications. Surgery is indicated either through hemispherectomy or corpus callosotomy for those with refractory seizures.13,14 Intraocular pressure monitoring allows early glaucoma detection and therapy initiation to prevent optic nerve damage. Some patients may require surgery, such as goniotomy or trabeculectomy. For the facial port wine stain, laser treatment can result in lightening of the port-wine birthmark therefore Pulsed dye laser (PDL) therapy should be started as soon as possible.15,16 Thus management of SWS requires a multidisciplinary approach that includes a dermatologist, a neurologist, and an ophthalmologist.

**Conclusion**

This case report highlighted the atypical feature of SWS, i.e. that is the contralateral brain involvement in the facial port wine stain. Research has shown that the prognosis of both ipsilateral and contralateral brain involvement in SWS is the same. The case also highlighted the importance of the multicenter approach in the treatment of SWS. Further research is needed to support the idea of contralateral brain involvement in SWS.

**Author's contribution:** AS was responsible for the conceptualization of this case report. AS, QAK, SYS, and SK were responsible for the clinical analysis. MJ was responsible for the image analysis. YLC, HP, VRG, and JN contributed to the preparation of the original draft. RV was responsible for the study supervision. All authors contributed to the writing of the final manuscript.

**Data availability statement:** Data can be available on reasonable request to the corresponding author.

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