ROACH'S TYPE II VARIANT OF STURGE – WEBER SYNDROME: A CASE REPORT

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ABSTRACT

Sturge- weber syndrome is a neurocutaneous disorder caused by persistence of transitory primordial arteriovenous connection of the foetal intracranial vasculature. It manifests with vascular malformations involving the brain, eye and skin with resulting neurological and orbital manifestations. Port wine stain, glaucoma and seizures are some of the commonly seen symptoms, depending on the presence of these features sturge weber syndrome has been classified by Roach into three types. We report a case of a 26 year old female with facial portwine stain and minimal neurologic manifestations, which according to Roach's classification falls in type II category. This report also underlines the need for detailed laboratory and neurologic work up of all patients with facial portwine stain present along the distribution of trigeminal nerve, as the neurologic manifestations in sturge–weber syndrome may vary in severity from seizures and mental retardation to minimal radiographic changes. It also emphasizes the need for identification of such rare variants of this syndrome.

ملخص: متلازمة ستيرج ويبر هو اضطراب عصبي جلدي ناجم عن استمرار اتصال الشرايين والأوردة الأولية المؤقتة داخل جمجمة الجنين، و يظهر في شكل تشوهات وعائية في المخ والعين والجلد و يؤدي الي أعراض عصبية وأعراض في محجر العين. من أعراضه الشائعة تلون الجلد باللون الاحمر القاني, ارتفاع ضغط العين وتشنجات. حسب الاعراض تم تصنيف متلازمة ستيرج ويبر من قبل روتش إلى ثلاثة أنواع. سجلنا حالة لأنثى تبلغ من العمر 26 عاما تشكو من تلون بجلد الوجه واعراض عصبية بسيطة والتي تصنف حسب روتش في الفئة الثانية. هذا التقرير يؤكد أيضا الحاجة إلى مزيد من التقصي السريري و المخبري لجميع المرضى الذين يعانون من تلون الوجه على طول مسار العصب الثلاثي التوائم ، وخصوصا ان الاعراض العصبية في متلازمة ستيرج ويبر قد تختلف في شدتها من التشنجات و التخلف العقلي الي التغيرات البسيطة في الفحص الشعاعي. التقرير يؤكد أيضا على الحاجة إلى تحديد المتغيرات النادرة لمثل هذه المتلازمة.

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INTRODUCTION

Sturge-Weber Syndrome also called as encephalotrigeminal angiomatosis is an uncommon condition that is characterized by hamartomatous vascular proliferation involving the tissues of the brain and face. It occurs due to persistence of vascular plexus around the cephalic portion of the neural tube. This plexus develops during the sixth week of intrauterine life and usually regresses by the ninth week. The exact cause is unknown but a "2-hit hypothesis" which involves sporadic mutations as well as familial occurrences has been suggested as etiological basis[1]. Most cases are sporadic but occasionally cases within families have also been reported. Males and females seem to be equally affected. It has been reported in individuals of White, Hispanic, African and Asian heritage.

Clinically, patients typically presents with constellation of signs and symptoms such as congenital facial Angiomas (Port Wine Stain/PWS)[2,3], glaucoma, and variable neurologic manifestations including seizures, mental retardation, hemianopia, hemiparesis and learning difficulties[4-6]. Patients may also have emotional problems, such as depression, low self-esteem, shame, emotional outbursts and isolation. The facial angioma is usually unilateral but may be bilateral. It typically involves at least the upper face, superior eyelid, or periorbital region. The facial angioma conforms to sensory distribution of the trigeminal nerve, Angiomas may also involve gingiva, nasopharynx, palate, lips and tongue[7,8]. Port wine stain may also be found on the trunk or extremities of some individuals with Sturge Weber Syndrome.

Seizures and other neurologic complications are the result of leptomeningeal angiomas. The severity of the neurological manifestations depends on the location and amount of area involved by leptomeningeal angioma.

Glaucoma is also commonly seen in Sturge Weber Syndrome and is present in 60% of individuals. It can be present at birth or occur anytime throughout the lifespan. It can be unilateral or bilateral, untreated, glaucoma can cause blindness and can be extremely painful.



Figure 1: Characteristic unilateral involvement of face with ophthalmic and maxillary nerves affected

Depending on the presence of facial angiomas, leptomeningeal angiomas and glaucoma, Roach has classified Sturge Weber Syndrome as follows[9]

Type I - Both facial and leptomeningeal angiomas; may have glaucoma

Type II - Facial angioma alone (no CNS involvement); may have glaucoma

Type III - Isolated LA; usually no glaucoma

CASE HISTORY

A 26 year lady reported to the outpatient department with complaint of facial nevi involving the upper face on right side, she also complained of bleeding from the

gums. On detailed history patient reported nevi to be congenital, initially with pink colour and later progressing to purplish hue. There was no history of seizure or mental retardation but during examination the patient was dull and was answering with difficulty. On examination, the nevi was extending superiorly from the periorbital region to the angle of the mouth inferiorly, upto the midline (Fig 1).

Table 1 Clinical Manifestations of Sturge-Weber Syndrome

Risk of SWS with facial PWS	8%
SWS without facial nevus	13%
Bilateral cerebral involvement	15%
Seizures	72-93%
Hemiparesis	25-56%
Hemianopia	44%
Headache	44-62%
Developmental delay and mental retardation	50-75%
Glaucoma	30-71%
Choroidal Haemangioma	40%

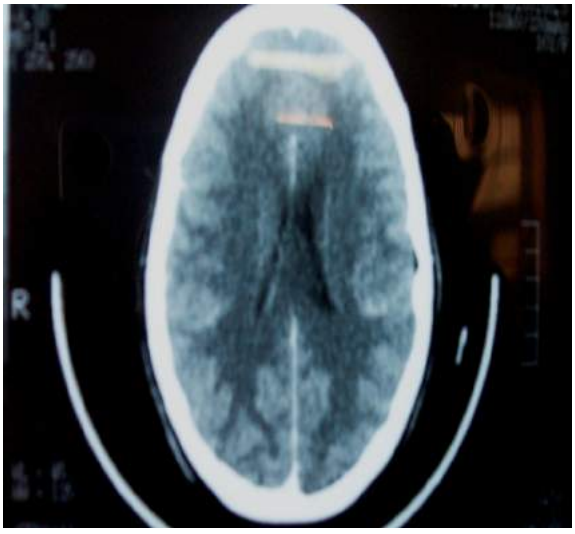


Figure 2: CT scan revealing mild cortical atrophy and focal calvarial thickening.

The CT scan revealed minor changes like gyriform enhancement in parietal lobe with mild cortical atrophy and focal calvarial thickening (Fig 2). Eye examination revealed no glaucoma and there was no history of pain or imperfect vision Intraorally the gingival hyperplasia was noticed in both upper and lower arch on right side characteristically extending till midline (Fig 3 and 4).



Figure 3: Hyperplastic maxillary gingiva extending till midline

Intra oral examination revealed gingival hyperplasia of both the arches extending up to the midline. Plaque and calculus was seen in the region of gingival enlargement, but this was because the patient could not brush that region due to bleeding. Gingival hyperplasia was more pronounced on the maxillary arch and covered almost the whole crown, buccal mucosa on the right side was also erythematous. The MRI findings revealed enhanced soft tissue mass lesion in the right oropharynx (Fig 5).



Figure 4: Gingival enlargement in lower arch is less compared to upper arch.

DISCUSSION

The Sturge-Weber syndrome (SWS) is a neurocutaneous disorder with angiomas involving the leptomeninges and skin of the face, typically in the ophthalmic and maxillary distributions of the trigeminal nerve. SWS is caused by residual embryonal blood vessels and their secondary effects on surrounding brain tissue. A vascular plexus develops around the cephalic portion of the neural tube, and normally regresses around the ninth week of gestation. Failure of this normal regression results in residual vascular tissue, which forms the angiomata of the leptomeninges, face, and ipsilateral eye. In a study by Tallman et al it was reported that 310 patients with PWS; 85% had unilateral and 15% had bilateral involvement, and 68% had involvement of more than 1 dermatome. Only patients with PWS involving the distributions of the V1 and V2 branches of the trigeminal nerve had CNS or eye involvement. Overall, in those with trigeminal involvement, only 8% had CNS and eye involvement[10]. Port wine stain is also associated with soft-tissue hypertrophy: The Sturge-Weber Foundation survey indicated that body asymmetry was seen in 164 of 171 patients, with soft-tissue hypertrophy in 38 of 164 patients and scoliosis in 11 patients. Basal cell carcinoma has been reported to occur within a PWS[11]. Intraorally, hypervascular changes may be seen on the ipsilateral mucosa and gingiva ranging from slight vascular hyperplasia to more massive hemangiomatous proliferation resembling a pyogenic granuloma.

Neurologic dysfunction results from secondary effects on surrounding brain tissue, which include hypoxia, ischemia, venous occlusion, thrombosis, infarction, or vasomotor phenomenon. A "vascular steal phenomenon"[12] may develop around the angioma, resulting in cortical ischemia and progressing to calcification, gliosis, and atrophy, which in turn increase the chance of seizures and neurologic deterioration[13]. The incidence of epilepsy in patients with SWS is 75-90%; seizures may be intractable. Seizures result from cortical irritability caused by cerebral angioma, through mechanisms of hypoxia, ischemia, and gliosis. Fibronectin is a molecule important in regulating angiogenesis, maintenance of the blood-brain barrier, blood vessel structure and function, as well as brain tissue responses to seizures. Comi et al reported that, in patients with SWS, decreased expression of fibronectin was noted in the leptomeningeal blood vessels[14].

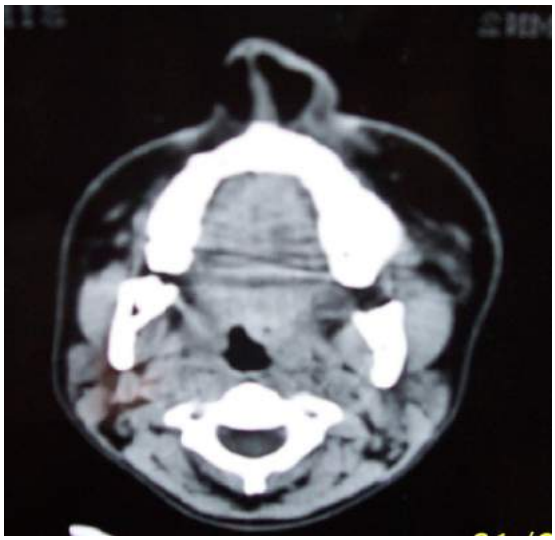


Figure 5: MRI revealing hyperplasia of right side

The main ocular manifestations (ie, buphthalmos, glaucoma) occur secondary to increased intra ocular pressure (IOP) with mechanical obstruction of the angle of the eye, elevated episcleral venous pressure, or increased secretion of aqueous fluid. Untreated, glaucoma can cause blindness. It can be extremely painful and may be a "silent" cause of behavior outbursts or self-injurious behavior in non-verbal individuals with Sturge Weber Syndrome. The incidences of the major

clinical manifestations of SWS are listed in Table 1[15].

In our case the patient complained only of the port wine stain and gingival hypertrophy with no history of seizure or ocular manifestations. The gingival hyperplasia reported in SWS has sometimes been attributed to the medications used for seizure control, but in our case the patient was not taking any medications and the hyperplasia was seen characteristically only upto the midline. The CT scan of brain did not show any obvious changes or calcifications but minor changes such as gyriform enhancement and some amount of cortical atrophy of the right half can be appreciated. This minimal involvement of the brain correlates well with the lack of neurologic manifestations seen in patient. This type of presentation of SWS is very rare and only reported by Bioxeda et al in 1993[16].

CONCLUSION

This is a rare case of SWS, which has minimal CNS involvement but no neurologic manifestations and hence this falls in Type II SWS according to Roach. Hence we suggest that any case having facial nevus along the course of trigeminal nerve should be suspected for SWS because the neurologic involvement in a given case may vary from minimal radiographic changes to overt clinical manifestations.

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