Case Reports

Yam Bahadur Roka
MS, MCh, IFAANS

Address for correspondence:
Dr. Yam Bahadur Roka
M.S. (Surgery), M.Ch. (Neurosurgery), F.N.N.I. (Skull Base Surgery), IFAANS
Chief of Neurosurgery
Neuro Cardio and Multispeciality Hospital
Biratnagar, Nepal
Email: dryamroka@yahoo.com

Date submitted : 10 July 2018
Date accepted : 15 August 2018

Sturge Weber syndrome

Sturge-weber syndrome is a type of neurocutaneous syndrome/neurooculo cutaneous/phakomatoses that is characterized by facial capillary Port Wine stain, leptomeningealangiomatosis and glaucoma with an incidence of 1 per 20-50,000 live births. A case is reported that presented to the outpatient clinic with discharging wound over the left side of his head for a week. On examination he had a large left facial PW stain involving all the divisions of the Trigeminal nerve. Further examination revealed he had history of partial seizures since last 10 years with incomplete and irregular treatment. He also had right sided hemiatrophy of the limbs along with medial gaze deviation of the left eyeball. Skull skiagram was done outside which revealed calcifications in his left hemisphere and CT was then advised for his headache and seizure that revealed Left hemispheric Tram track calcifications along with cerebral atrophy and loss of cortical volume. EEG was unremarkable. He was managed with antibiotics for the scalp wound and discharged with Leveteracetam 1000 mg daily dose and asked to attend eye hospital for the reduced vision. With the PW stain, skin nodules, eye findings and the radiological features he was diagnosed as SWS and counseled for regular follow up, use of anti-epileptic medications, probable side effects and surgery for eye abnormalities.

Keywords: Glaucoma, Port Wine Stain, Sturge Weber Syndrome, Neurocutaneous Syndrome, NeuroOculo Cutaneous, Phakomatoses, Seizure,
three types (Table 1). SWS may manifest with cutaneous, neurological, endocrine or ocular manifestations. An interesting case of SWS is reported with the classical findings.

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Roach I</td>
<td>facial PWS and lepto meningioma, with or without associated glaucoma, consistent to typical SWS</td>
</tr>
<tr>
<td>Roach II</td>
<td>more common, with facial PWS and no lepto meningioma involving, with or without presence of glaucoma</td>
</tr>
<tr>
<td>Roach III</td>
<td>which is the least frequent form, with presence only of lepto meningioma</td>
</tr>
</tbody>
</table>

Table 1. The three types of Roach SWS scale and their manifestations.

**Case Report**

An 18-year male from India presented to the outpatient clinic with discharging wound over the left side of his head for a week. He had history of two lumps over the scalp since birth and the larger had been partially excised leading to bleeding and secondary infection. On examination he was conscious and orientated and had a large left facial PW stain involving all the divisions of the Trigeminal nerve. Further examination revealed he had history of partial seizures since last 10 years with incomplete and irregular treatment. He also had episodic migraine like headaches on his left side. There was also history of reduced vision for the past 2 years in his left eye and had been taking eye drops from a local practitioner. He also had right sided hemiatrophy of the limbs along with medial gaze deviation of the left eyeball.

Skull skiagram was done outside which revealed calcifications in his left hemisphere and CT was then advised for his headache and seizure that revealed left hemispheric Tram track calcifications along with cerebral atrophy and loss of cortical volume. EEG was unremarkable. He was managed with antibiotics for the scalp wound and discharged with Leveteracetam 1000 mg daily dose and asked to attend eye hospital for the reduced vision. With the PW stain, skin nodules, eye findings and the radiological features he was diagnosed as SWS and the family counseled regarding the disease. He was also counseled for regular follow up, use of anti-epileptic medications, probable side effects and surgery for eye abnormalities.

**Discussion:**

SWS are an uncommon disorder of the vascular supply of the head causing neuro oculocutaneous symptoms. The other names for this condition are...
“angiomatos is aculoorbital-thalamic syndrome, meningo- 
oculo-facial angiomatosis, neuroretinoangiomaticosis, 
encephal of acialhemangiomaticosis syndrome, mening 
of acialangiomatos is-cerebral calci 
ciffation syndrome, 
Sturge-Weber-Dimitri syndrome phakomatosis, encephal 
of acialhemangiomaticosis, and Sturge-Weber-Krabbe 
syndrome”.6It is a somatic mutation and hence not 
inherited. The mosaicism during early development 
leads to abnormal vessel formation is some parts of the 
brain only. The PS stain is usually present at birth and 
the clinical symptoms may manifest after 2 years of age. 
The abnormal vasculature leads to formation of LA, 
calcifications and cerebral atrophy on the affected side. 
This can lead to temporary stroke like symptoms,seizures, 
 transient hemiparesis or vision loss, headaches and 
migraine like symptoms.

Neurological features: SWS can involve ipsilateral 
or bilateral, parietal, occipital, parieto-occipital or 
complete hemispheric area with LA. They are made of 
abnormal tortuous vessels with associated atrophy of the 
ipsilateral cortex, cortical dysgenesis, calcifications and 
cerebral atrophy. They present with seizures (75-90%), 
slow progressive hemiparesis (25-60%), infantile spasm, 
mental retardation, migraine like vascular headaches (30 
-40%), delayed milestones (50-60%), behavioral or social 
problems and visual field defects.3,3,5

Cutaneous features: the classic picture is that of PW 
stain which is asymmetrically located on the face and may 
be unilateral or bilateral. They are mostly seen at birth 
and needs to be differentiated from salmon patches- nevus simplex. PW stain may affect any of the three cutaneous 
divisions of the trigeminal nerve and those associated 
with V1 bilateral or extensive involvement of frontal skin, 
have higher tendency for PW-LA complex formation or 
Glaucoma. PW may lead to local facial tissue hypertrophy, 
hepi atrophy of the face, bone or maxillary overgrowth, 
jaw malocclusion or formation of nodules.5,9

Ocular features:dilated vessels that involve the 
choroid, retina, or conjunctiva are the hall mark of 
SWS. This can lead to glaucoma, optic atrophy or even 
blindness. Choroidalhemangiomas are present in 40-50% 
of cases and seen as tomato catsup color. Glaucoma on the 
same side of the lesion can occur in as many as 70% of 
cases and is secondary to the abnormal anterior chamber 
angle or increase in episcleral venous pressure anomalies 
 obstructing the flow of aqueous humor. The other findings 
that are associated with SWS are strabismus, enlarged 
eyeball (buphthalmos), lens luxation, retinal detachment, 
heterochromia and homonymous hemianopsia.3,10 EDI 
SD-OCT can show choroidal thickness above 1000 m and 
B-Scan will show the diffuse choroidal hemangioma.4,10

Figure 2. CT head showing extensive intracranial left hemispheric calcification with cerebral 
atrophy. There are no abnormal dilated venous channels on contrast scan.
**Endocrine features:** hypothyroidism with growth hormone deficiency may be associated.  

**Diagnosis:** is by clinical and radiological investigations. Electro encephalogram is usually done to classify the type of seizure. Plain Skiagram will show the classic rail road track calcifications which are in the parieto-occipital lobe or involve the ipsilateral hemisphere. CT will show similar findings to that of the Skiagram but more detailed, detecting calcifications at an early age, and the extent of the lesion, the hemisphere involved and the nature of the cortex will be better outlined. MRI with contrast remains the most important method of investigation. It can detect the calcifications, the extent of cerebral atrophy, the nature and pattern of the abnormal vessels with the enlarged choroid plexus. It is usually advised after first year of life. MRI will show temporary hyperperfusion with leptomeningeal enhancement (serpiginous) and in the late phase, increased T2 signal in the area of gliosis with decreased pial enhancement and cortical atrophy. Cerebral angiography is not routinely done and if indicated can help distinguish calcifications from arteriovenous malformations or to define the venous drainage. The clinical findings supported with the radiological characteristics will help clinch the diagnosis of SWS. PW stain with eye findings is sufficient for diagnosis and there is a debate whether further investigation has to be done for confirmation.

**Treatment:** consists of mainly family counseling regarding the disease along with medical management for control of seizures, headache, glaucoma and cosmetic therapy (Laser therapy) for the PW stain. The patient should be on regular follow up with dose adjustment according to weight and frequent check up to rule out the adverse effects of prolonged anti-convulsion medications. Aspirin for transient hemiparesis or Flunarizine for associated migraine is advised. For medically refractory seizures surgery may be indicated in the form of lesionectomy, corpus callosotomy or even hemispherectomy. Glaucoma can also be managed with surgery but has poorer results in comparison with primary angle glaucoma. Retinal hemorrhage may occur due to the sudden lowering of intraocular pressure during surgery. Simultaneous monitoring of the hormonal profile is also needed. The prognosis of SWS is usually good. Early age seizure, extensive cerebral or ocular involvements are poor prognosis factors.

**Conclusion**

SWS are one of the neurocutaneous syndromes that can present to the outpatient department. Clinical findings, EEG and CT or MRI are sufficient for its diagnosis. Management requires use of anti-epileptic medications along with glaucoma care.

**Sturge Weber Syndrome**

**References**