**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

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# Sturge Weber syndrome

Sturge-weber syndrome is a type of neurocutaneous syndrome/ neurooculo cutaneous/ phakomatoses that is characterized by facial capillary Port Wine stain, leptomeningealangioma and glaucoma with a 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 revealedLeft 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:**Galucoma,PortWineStain,Sturge Weber Syndrome, Neurocutaneous Syndrome, NeuroOculo Cutaneous, Phakomatoses, Seizure,

Sturge-weber syndrome (SWS) is a type of neurocutaneous syndrome/ neurooculo cutaneous/ phakomatoses that is characterized by facial capillary Port Wine stain, leptomeningealangioma and glaucoma with a incidence of 1 per 20-50,000 live births.[4,5]Somatic mutation in the GNAQ gene located on chromosome 9 affects the G proteins that act as modulators/translators in transmembrane signaling systems and is found in 80% of cases with SWS and the dysregulation of vascular MAPK and/or PI3K signaling during human embryonic development leads of SWS.[8] The port wine stain and the leptomeningealangiomatosis ( PW-LA) are due to the failure of the cutaneous venous plexus to regress. Normally the primitive venous system is comprised of three types, the external skin, the middle leptomeningeal and the internal brain systems that drain their respective areas. The upper part of the facial skin is located near to the future parieto-occipital area of the brain and thus mutations of this region affect both the areas. It has been classically described by the Roach scale into

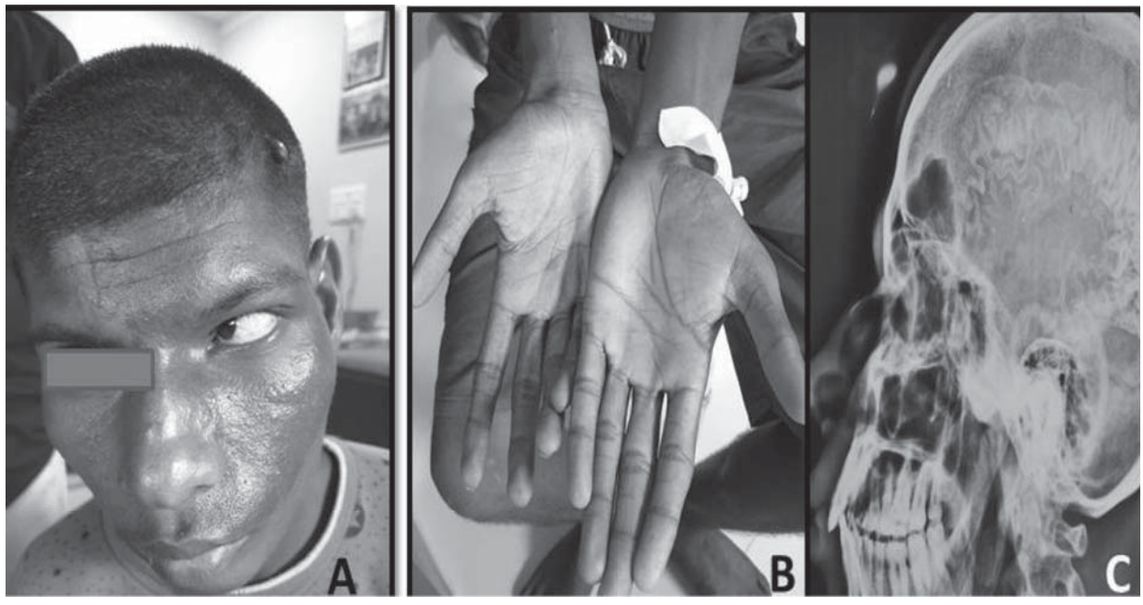


*Figure 1.The PW satin on the left side of the face involving all three divisions of trigeminal nerve with left VI th nerve palsy. There were multiple scalp anngiomatous formations on left side (A). Note the associated right sided hemiatrophy (B). Skull skiagram showing extensive intracranial calcifications (C).*

three types(Table 1).[5] SWS may manifest with cutaneous, neurological, endocrine or ocular manifestations. An interesting case of SWS is reported with the classical findings.

| Type | Description |
|---|---|
| Roach I | facial PWS and lepto mening ealangioma tosis, with or without associated glaucoma, consistent to typical SWS |
| Roach II | more common, with facial PWS and no lepto mening ealinvolvement, with or without presence of glaucoma |
| Roach III | which is the least frequent form, with presence only of lepto mening ealangiomatosis |

*Table 1. The three types of Roach SWS scale and their manifestations.*[5]

## 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 revealedLeft 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 arean uncommon disorder of the vascular supply of the head causing neuro oculocutaneous symptoms. The other names for this condition are

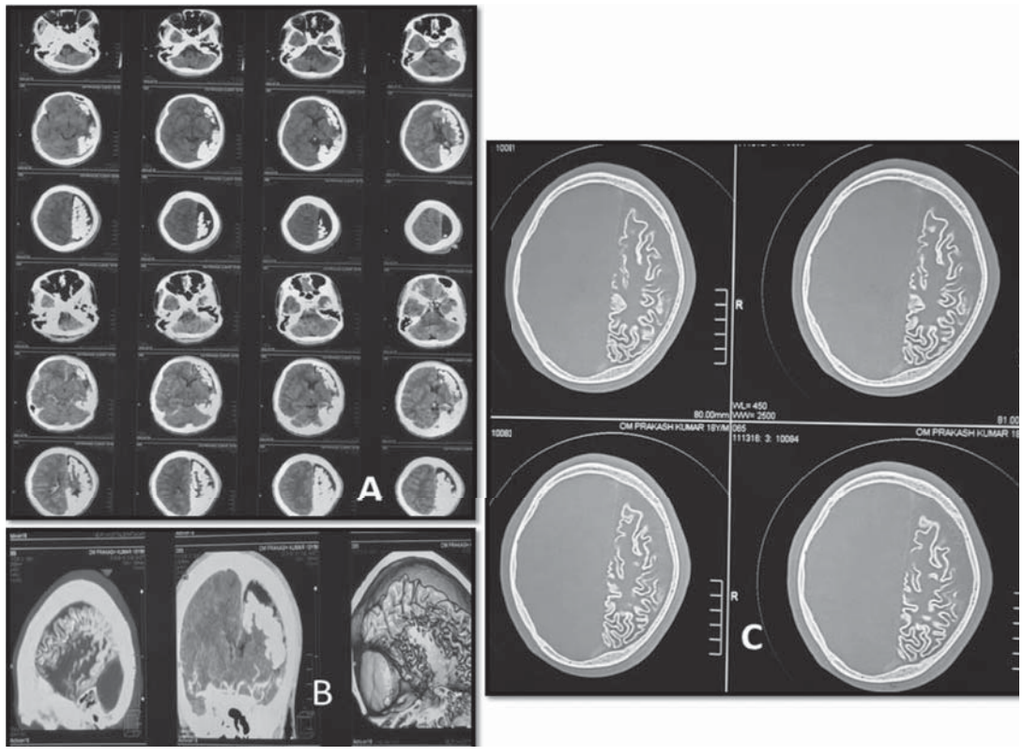


*Figure 2.CT head showing extensive intracranial left hemispheric calcification with cerebral atrophy. There are no abnormal dilated venous channels on contrast scan.*

"angiomatos is aculoorbital-thalamic syndrome, meningo-oculo-facial angiomatosis, neuroretinoangiomatosis, encephal of acialhemangiomatosis syndrome, mening of acialangiomatos is-cerebral calcification syndrome, Sturge-Weber-Dimitri syndrome phakomatosis, encephal of acialhemangiomatosis, 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.[2,3,5,7]

**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, hemi 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 andis 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 (bupthalmos), lens luxation, retinal detachment, heterochromia and homonymous hemianopsia.[5,10] EDI SD-OCT can show choroidal thickness above 1000 □m and B-Scan will show the diffuse choroidal hemangioma.[4,10]

**Endocrine features:** hypothyroidism with growth hormone deficiency may be associated.[1]

**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.[1,7]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.[11]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.[5,12] 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.[3]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 neurocutaneoussyndromes 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.

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