

Sturge-Weber syndrome type III: an important stroke mimic

Sumita Manohari,¹ Pawan Banyal,¹ Arushi Gahlot Saini ,¹ Sameer Vyas²

¹Pediatrics, PGIMER, Chandigarh, Chandigarh, India
²Radiodiagnosis, Post Graduate Institute of Medical Education and Research, Chandigarh, Chandigarh, India

Correspondence to
Dr Arushi Gahlot Saini;
doc.arushi@gmail.com

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DESCRIPTION

A girl in middle childhood presented with prolonged right-sided tonic seizures with focal status, and right hemiparesis. She had a seizure with transient right-sided weakness 1 week before but had recovered completely. Birth and perinatal period were normal. She had one generalised seizure at 1 year of age for which antiseizure medications were required. The treatment details of this time were not available. Family history was unremarkable. On examination, she was conscious and oriented with normal speech with right hemiplegia and right-sided upper motor neuron type of facial nerve palsy. Deep tendon reflexes were absent on the right side and power at proximal and distal joints was 1/5. There were no neurocutaneous markers. Rest of the systemic examination was normal. An initial clinical diagnosis of right-sided ischaemic stroke was considered. CT head showed multiple cortical calcifications in the left cerebral hemispheres (**figure 1A–B**). Skull radiograph demonstrated the tram-track calcification. MRI of the brain revealed pial angiomatosis and calcifications along gyri in left cerebral hemisphere with mild cerebral hemiatrophy (**figure 1C–H**). EEG showed generalised delta theta slowing and asymmetric right background activity and voltage attenuation suggestive of diffuse (right > left) cortical dysfunction (**figure 2**). A clinico-radiological diagnosis of Sturge-Weber syndrome

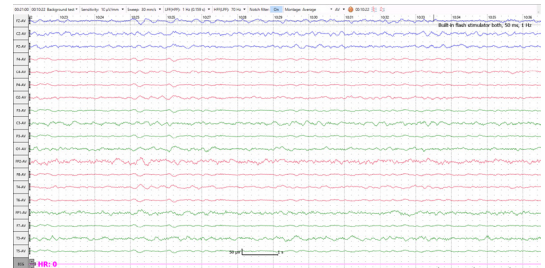


Figure 2 Sedated sleep EEG record obtained with 10–20 system of electrode placement shows generalised delta theta slowing with paucity of sleep markers and asymmetric right background activity and voltage attenuation suggestive of diffuse (right>left) cortical dysfunction.

type III was considered. She was continued on anti-seizure medications drugs and oral aspirin (5 mg/kg/day).

SWS is the third most common neurocutaneous disorder after neurofibromatosis and tuberous sclerosis complex. It is a rare, congenital, non-hereditary, developmental disorder where hamartomatous vascular proliferation occurs in the tissues of brain, face and eyes that leads to complications related to the site and extent of involvement.¹ It is often classified into

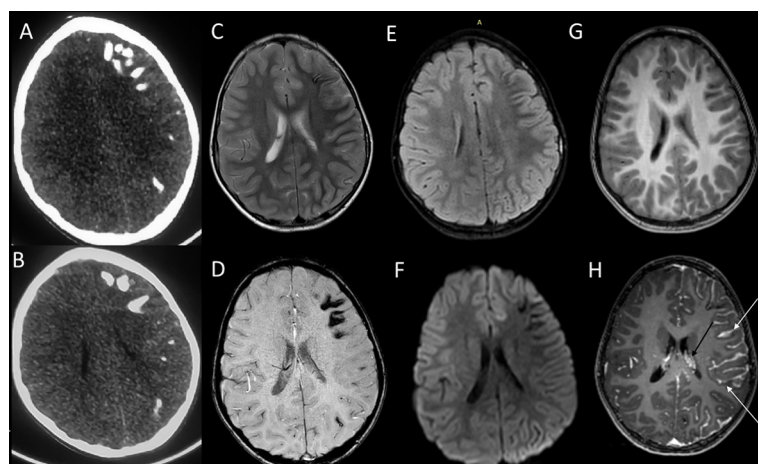


Figure 1 (A–B) CT head axial section showing extensive gyral and subcortical calcification at the left frontal and parietal regions of left cerebral hemisphere. (C) Axial T2-weighted, (D) Fluid-attenuated inversion recovery (FLAIR), (E) T1-weighted, (F) susceptibility-weighted (SWI), (G) diffusion-weighted ($b=1000\text{ s/mm}^2$) and (H) contrast-enhanced T1-weighted MR images showing mildly prominent extra-axial CSF spaces in left cerebral hemisphere. Cortical T2-hypointensity is seen in left frontal lobe (C) with marked susceptibility changes on SWI images (F). There is marked leptomeningeal enhancement on left side (white arrows in H) with prominent choroid plexus (black arrow). Note made of diffuse hypointensity in left cerebral white matter on T2-weighted and FLAIR images, more marked in subcortical white matter likely due to seizures or ischaemia.



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three main types: type I with skin and neurological symptoms with or without glaucoma, type II with only skin and no neurological involvement with possible glaucoma and type III with isolated neurological involvement without skin abnormality and usually no glaucoma.² Somatic mutation of the *GNAQ* gene is implicated in pathogenesis, which causes persistence of vascular plexus around the cephalic portion of the neural tube that fails to regress during the ninth week of gestation and in later course of life undergoes pathological remodelling, venous stasis and chronic ischaemia.² Proteins and calcium ions entering cells through increased vascular permeability in these capillaries cause crystallisation and calcification, which help in early diagnosis on neuroimaging.

The typical skin lesions 'port-wine stain' presents as a birthmark in the distribution of first and second divisions of trigeminal nerve. However, it may be absent in 13% of cases.² In such cases, the neurological presentation often mimics an acute or recurrent ischaemic stroke secondary to an arteriopathy or thromboembolic event. Missed cases may present later with refractory epilepsy (75–90%), intellectual impairment (50%) or hemiplegia/hemiatrophy (30%).³ The cortical leptomeningeal angiomas causes atrophy, neuronal loss and astrogliosis in the underlying brain tissue resulting in structural alterations. During a seizure in such a patient, instead of normal response of increased blood flow, there is rather a decrease in the blood flow to ischaemic levels, which contributes to stroke and stroke-like episodes. Such changes can be measured by perfusion MRI sequences. Gadolinium-enhanced T1-weighted and susceptibility-weighted MRI are more sensitive than CT for early detection of the leptomeningeal capillary-venous malformation. Thus, it is important to recognise SWS type III as an important stroke mimic in the emergency room. There is increasing agreement that low-dose aspirin (3–5 mg/kg/day) as an antithrombotic therapy be started in children with SWS beginning in infancy to reduce the frequency of strokes and stroke-like events, as well as seizures.¹ In the absence of controlled studies, the evidence is currently based on observational studies only.^{4 5}

Learning points

- ▶ Sturge-Weber syndrome (SWS) is the third most common neurocutaneous disorder after neurofibromatosis and tuberous sclerosis complex.
- ▶ SWS type III as an important stroke mimic.
- ▶ Gadolinium-enhanced T1-weighted and susceptibility-weighted MRI are more sensitive than CT for early detection of the leptomeningeal capillary-venous malformation.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

ORCID iD

Arushi Gahlot Saini <http://orcid.org/0000-0002-5747-8975>

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