CASE REPORT

Bilateral intracranial calcifications with bilateral facial cutaneous naevus: Sturge Weber syndrome

Muhammad Arif Saeed,¹ Kiran Hilal,¹ Prem Chand²¹Department of Radiology, Aga Khan University, Karachi, Pakistan²Department of Pediatrics and Child Health, Aga Khan University Hospital, Karachi, Pakistan**Correspondence to**

Dr Muhammad Arif Saeed, arifsaeed10@hotmail.com, noman_452@hotmail.com

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SUMMARY

Sturge Weber syndrome also known as mother spot disease belongs to a group of disorders called phakomatoses (neurocutaneous syndromes involving the central nervous and cutaneous systems). It is a congenital disorder of the vasculature of the meninges, brain, face and eyes. Clinically, it usually presents with seizures and other neurological complications, including mental retardation, contralateral hemiparesis and glaucoma. Its incidence is estimated at one case in 20 000–50 000 persons with equal frequency in boys and girls. We present a case of 18-months-old boy was brought by parents with history of seizures since birth resulting in fall 1 day back. Physical examination showed bilateral port-wine stain on face. CT head examination revealed bilateral intracranial calcification. This is a highly unusual presentation of Sturge Weber syndrome.

BACKGROUND

Sturge Weber syndrome¹ (SWS) is a rare neurocutaneous syndrome characterised by facial port-wine stain involving the first division of the trigeminal nerve, ipsilateral leptomeningeal angiomas and angioma involving the ipsilateral eye.² It is also known as encephalotrigeminal

angiomatosis. We report a radiologically atypical presentation of this syndrome.

CASE PRESENTATION

An 18-month-old boy was brought by parents with history of seizures resulting in fall 1 day back. There was no history of neonatal asphyxia or hypoglycaemia. There was no family history of epilepsy. On examination, the patient was awake and alert, with no bruise at site of fall. Pupils were bilaterally equal in size and reactive to light. There was no asymmetry in movements in all limbs. No aphasia was noted. On physical examination, the patient had bilateral facial redness consistent with bilateral port-wine stain shown in [figure 1](#). There was no positive finding on systemic examination. All examination was normal.

INVESTIGATIONS

All the biochemical test results were within normal limits:

serum glucose: 90 mg/dL
total bilirubin: 0.9 mg/dL
haemoglobin: 12 g/dL
white cell count: $7 \times 10^9/L$
alanine transaminase: 45 IU/dL
serum creatinine: 0.9 mg/dL.

RADIOLOGICAL INVESTIGATIONS

In keeping with the facial port-wine stain and history of seizures, further radiological workup was done. Non-contrast CT scan of head was conducted in the axial plane with sagittal and coronal reconstruction using Toshiba Aquilon 64 slice machine. It revealed subcortical calcifications in the right occipital and left temporal lobes shown in [figures 2–5](#).

MRI brain with contrast was done which revealed abnormal hyperintense signals along the gyri and the cerebral convexity in the left temporal ([figures 6 and 7](#)) and right occipital lobe ([figures 8 and 9](#)) with corresponding area of post-contrast enhancement. Bilateral choroid plexuses appear prominent. Mild diffuse leptomeningeal thickening and enhancement was also noted. The clinical history and radiological findings were consistent with diagnosis of Type I SWS with bilateral calcifications, an extremely uncommon finding.

Figure 1 Bilateral naevus.

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Figure 2 Unenhanced CT head axial section showing right occipital calcification.

OUTCOME AND FOLLOW-UP

Patient was started on phenobarbital orally for seizure control and was followed. He remained seizure-free to the time of publication.

DISCUSSION

Intracranial calcification and facial naevus is the characteristic of SWS. It is usually seen unilaterally. Bilateral intracranial calcification is seen in approximately 15% of patients and portends an unfavourable prognosis.³ Port-wine stain is the most common clinical manifestation of SWS, usually presenting unilaterally, typically on the forehead and upper eyelid, and it may be extended to the neck and other parts of

Figure 4 Unenhanced CT head axial section showing left temporal calcification.

the body.⁴ Ischaemic changes in the tissue surrounding the lesion with unilateral occipital lobe gyriform calcification lead to convulsions, hemiparesis and cognitive dysfunction.⁵ Although bilateral naevus and bilateral intracranial calcifications in SWS have separately been described, simultaneous presentation has been extremely rare. Its early presentation with seizures in less than 2 year of age is a poor prognostic indicator. Prior studies have shown that bilateral naevus is of no help in predicting bilateral intracranial calcification since all patients who have bilateral port-wine stain do not have bilateral intracranial calcifications. Intracranial calcifications correlate with earlier onset of seizure. This case

Figure 3 Unenhanced CT head coronal section showing right occipital calcification.

Figure 5 Unenhanced CT head coronal section showing left temporal calcification.

Figure 6 MRI brain axial T1 weighted postcontrast abnormal signals along the gyri and the cerebral convexity in the right occipital lobe.

Figure 8 MRI brain axial T1 weighted postcontrast abnormal signals along the gyri and the cerebral convexity in the left temporal lobe.

Figure 7 Coronal fluid-attenuated inversion recovery postcontrast abnormal signals along the gyri and the cerebral convexity in the right occipital lobe.

Figure 9 Coronal fluid-attenuated inversion recovery postcontrast abnormal signals along the gyri and the cerebral convexity in the left temporal lobe.

Learning points

- ▶ Sturge Weber syndrome can have bilateral intracranial calcifications.
- ▶ Patient with port-wine stain should be screened for Sturge Weber syndrome.
- ▶ A patient who has bilateral port-wine stain can have bilateral intracranial calcifications.

is unique as the patient had bilateral naevus with bilateral intracranial calcifications. All patients with facial naevus should undergo radiological evaluation to rule out SWS.

Contributors MAS obtained informed consent from the family and drafted the initial manuscript. KH reported CT and MRI scans of the patient and revised initial manuscript. PC carried out physical examination and clinical management of the patient.

Competing interests None declared.

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