International Journal of Radiology Sciences

ISSN Print: 2664-9810
ISSN Online: 2664-9829
IJRC 2025; 7(2): 29-32
www.radiologyjournals.com
Received: 14-05-2025
Accepted: 19-06-2025

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Intractable epilepsy and crossed cerebellar diaschisis in pediatric sturge-weber syndrome: A case report

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DOI: https://doi.org/10.33545/26649810.2025.v7.i2a.39

Abstract
Sturge-Weber syndrome (SWS) is a rare neurocutaneous disorder characterized by facial port-wine stains, leptomeningeal angiomatosis, and ocular abnormalities. We present a 7-year-old girl with intractable seizures whose neuroimaging revealed classic features of SWS along with an unusual finding of crossed cerebellar diaschisis. Initial CT demonstrated right parieto-occipital and frontal tram-track calcifications with cerebral volume loss. MRI revealed right cerebral leptomeningeal angiomatosis with prominent medullary veins, choroid plexus enlargement, and notably, left cerebellar volume loss with signal abnormalities consistent with crossed cerebellar diaschisis. This finding, rarely reported in SWS, likely represents functional disconnection between the affected cerebral cortex and contralateral cerebellum due to chronic hypoperfusion. While infratentorial involvement in SWS is increasingly recognized, crossed cerebellar diaschisis remains an exceptional finding that may indicate more severe hemispheric dysfunction. Recognition of this phenomenon expands our understanding of SWS's neurological impact and may have prognostic implications for patient management

Keywords: Sturge-Weber syndrome, crossed cerebellar diaschisis, leptomeningeal angiomatosis, pediatric neuroimaging, tram-track calcifications, neurocutaneous disorders

Introduction
Sturge-Weber syndrome (SWS) is a sporadic neurocutaneous disorder affecting approximately 1 in 20,000-50,000 live births, characterized by the classic triad of facial port-wine stain, leptomeningeal angiomatosis, and glaucoma [1, 2]. The syndrome results from a somatic activating mutation in the GNAQ gene, leading to abnormal persistence of the primitive vascular plexus during embryonic development [3].
The neurological manifestations of SWS are primarily attributed to chronic venous stasis and hypoxia resulting from impaired cortical venous drainage. This leads to progressive calcification, atrophy, and gliosis of the affected brain regions [4]. While supratentorial involvement is typical, recent studies have highlighted that infratentorial structures may be affected in up to 11% of cases [5].
We present an unusual case of a 7-year-old girl with SWS who demonstrated not only the classic supratentorial findings but also crossed cerebellar diaschisis—a phenomenon rarely described in this condition. This finding expands our understanding of the neurological sequelae of SWS and may have important implications for prognosis and management.

Case Presentation
A 7-year-old right-handed girl presented to our pediatric neurology clinic with a history of medically refractory seizures that began at 18 months of age. Her mother reported that the seizures initially manifested as focal motor seizures affecting the left arm, progressing over the years to include secondary generalization despite trials of multiple antiepileptic medications, including levetiracetam, oxcarbazepine, and valproic acid.
The child was born at full term via an uncomplicated vaginal delivery with no perinatal complications. A facial port-wine stain involving the right forehead and upper eyelid was noted at birth, prompting early ophthalmologic evaluation that revealed ipsilateral glaucoma at 6 months of age. Developmental milestones were initially normal until seizure onset, after which mild left-sided weakness and cognitive delays became apparent.

On physical examination, the child was alert but showed mild developmental delay for her age. A prominent port-wine stain was visible over the right forehead and periorbital region, consistent with V1 distribution of the trigeminal nerve. Neurological examination revealed left hemiparesis with increased deep tendon reflexes and an extensor plantar response on the left side. Visual field testing demonstrated a left homonymous hemianopia.

Neuroimaging Findings

Initial non-contrast CT of the head (Figure 1A) demonstrated characteristic tram-track calcifications in the right parieto-occipital and frontal regions, involving both gyral and subcortical areas. Associated findings included right cerebral hemisphere volume loss and compensatory calvarial thickening on the affected side.

MRI of the brain provided more detailed characterization of the pathology:

- **Structural abnormalities:** T2-weighted and FLAIR sequences revealed diffuse right cerebral hemispheric atrophy with abnormal white matter signal hyperintensity in the right parieto-occipital region, suggesting dysmyelination or gliosis (Figure 1B).
- **Vascular abnormalities:** Post-gadolinium T1-weighted images (Figure 1D) demonstrated extensive leptomeningeal enhancement over the right cerebral hemisphere, particularly prominent in the parieto-occipital region. Bilateral midbrain and pons pial enhancement was noted (Figure 1E). Numerous enlarged medullary and deep draining veins were visible. The right choroid plexus was markedly enlarged and avidly enhancing. A small scalp hemangioma was noted in the right frontal region (Figure 1F).
- **Crossed cerebellar diaschisis:** Unexpectedly, T2-weighted and FLAIR images (Figure 2A, 2B and 2C)) revealed volume loss and patchy hyperintense signal abnormalities in the left cerebellar hemisphere, contralateral to the supratentorial abnormalities. No cerebellar enhancement or calcification was identified.
- **Advanced imaging:** Susceptibility-weighted imaging (SWI) confirmed extensive calcification appearing as areas of marked signal dropout in the right cerebral hemisphere (Figure 1C). MR perfusion imaging revealed decreased cerebral blood flow in the affected right hemisphere, with a prolonged mean transit time.

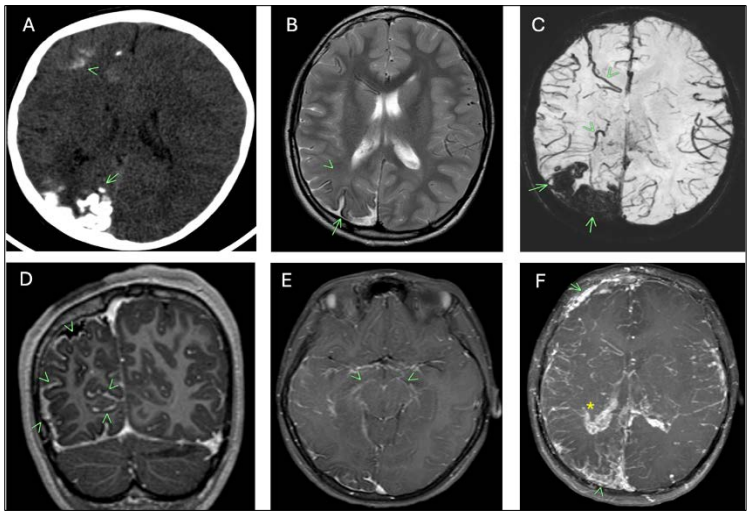

Fig 1: Neuroimaging findings of Sturge-Weber syndrome. (A) Axial non-contrast CT head demonstrated right cerebral atrophy and characteristic tram-track calcifications in the right parieto-occipital and frontal regions, involving both gyral and subcortical areas(*arrowheads*). (B) Axial T2-weighted demonstrates diffuse right cerebral hemispheric atrophy (arrow) with abnormal white matter signal hyperintensity (arrowhead) in the right parieto-occipital region, suggesting dysmyelination or gliosis. (C) Susceptibility-weighted imaging (SWI) confirmed extensive calcification appearing as areas of marked signal dropout in the right parieto-occipital region (*arrow*). Note is made of prominent medullary and deep draining veins in the right cerebral hemisphere (*arrowhead*). Post-contrast T1-weighted images in the coronal plane (D) and axial plane (E) demonstrated extensive leptomeningeal enhancement over the right cerebral hemisphere, particularly prominent in the parieto-occipital region (*arrowheads).* Bilateral midbrain and pons pial enhancement was noted (*arrowheads*). (F) Axial post-contrast T1-weighted MIP images demonstrate extensive leptomeningeal enhancement over the right cerebral hemisphere. Additionally, the right choroid plexus is enlarged and shows enhancement. There is a small scalp hemangioma in the right frontal region.

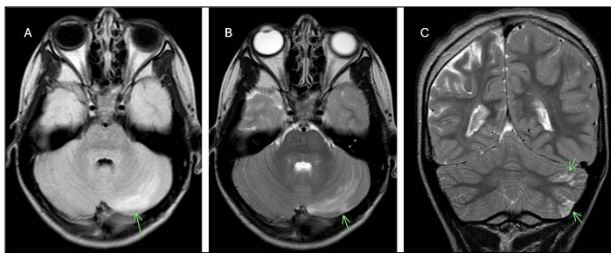

Fig 2: Crossed cerebellar diaschisis (A) Axial FLAIR image, T2-weighted image in axial(B)and coronal(C) planes demonstrate volume loss and patchy hyperintense signal abnormalities in the left cerebellar hemisphere, contralateral to the supratentorial abnormalities.

Discussion

This case illustrates the typical neuroimaging features of SWS while highlighting an unusual finding—crossed cerebellar diaschisis (CCD). The classic imaging findings in our patient, including tram-track calcifications, leptomeningeal angiomatosis, and enlarged ipsilateral choroid plexus, are well-established markers of SWS [6,7].

The pathophysiology of SWS centers on the persistence of primitive leptomeningeal vessels that fail to involute during embryonic development. This results in impaired cortical venous drainage, leading to chronic venous congestion, hypoxia, and ultimately neuronal loss with dystrophic calcification [8]. The characteristic tram-track pattern of

calcification follows the cortical gyri and represents both cortical and subcortical calcium deposition [9].

The finding of crossed cerebellar diaschisis in our patient represents a particularly interesting aspect of this case. CCD refers to the functional depression of the cerebellar hemisphere contralateral to a supratentorial lesion, mediated through disruption of the corticopontocerebellar pathways [10]. While CCD is well-documented in stroke, tumor, and epilepsy, it has been rarely reported in SWS [11].

The mechanism of CCD in SWS likely relates to chronic hypoperfusion and functional disconnection of the affected cerebral cortex from its cerebellar connections. Crossed cerebellar hypometabolism has been demonstrated on PET imaging in SWS patients, although structural changes, as seen in our case, are less commonly reported [12]. The presence of CCD may indicate more severe hemispheric dysfunction and could have prognostic implications for neurological outcome.

Recent literature has expanded our understanding of infratentorial involvement in SWS. Adams *et al*. reported that 11% of suspected SWS cases showed infratentorial pial angiomatosis, though isolated CCD without direct cerebellar angiomatosis, as in our case, remains exceptional [5]. This finding suggests that the neurological impact of SWS extends beyond direct vascular involvement to include remote functional effects.

The management implications of recognizing CCD in SWS are still being elucidated. Some authors suggest that the presence of CCD may indicate a higher seizure burden and poorer neurocognitive outcomes [13]. In our patient, the presence of intractable seizures despite multiple antiepileptic drugs and the development of hemiparesis support this association.

Treatment of SWS remains primarily supportive, focusing on seizure control, management of glaucoma, and rehabilitation for neurological deficits. Prophylactic anticonvulsants are increasingly advocated even before seizure onset [14]. Low-dose aspirin has been proposed to prevent stroke-like episodes by reducing venous thrombosis risk [15]. In cases of medically refractory epilepsy, as in our patient, surgical options including hemispherectomy may be considered [16].

Conclusions

This case illustrates that Sturge-Weber syndrome can present with crossed cerebellar diaschisis, a finding that expands the spectrum of neuroimaging abnormalities associated with this condition. Recognition of CCD in SWS patients may indicate more severe hemispheric dysfunction and could have important prognostic implications. As neuroimaging techniques continue to advance, subtle findings like CCD may help stratify patients for more aggressive management strategies. Further research is needed to determine the prevalence and clinical significance of crossed cerebellar diaschisis in the natural history of Sturge-Weber syndrome.

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