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Case Report

Refractory focal epilepsy in a child with Sturge–Weber syndrome: a case report

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ABSTRACT

Sturge–Weber syndrome (SWS) is a rare neurocutaneous disorder characterized by leptomeningeal angiomatosis, glaucoma, and port-wine nevus, frequently associated with refractory epilepsy and developmental delay. We report a 2-year-3-month-old boy with known SWS who presented with breakthrough focal motor seizures despite polytherapy. Clinical assessment revealed focal neurological deficits and global developmental delay. Neuroimaging demonstrated left cerebral atrophy with serpentine calcifications and choroid plexus enlargement consistent with SWS. Antiepileptic therapy optimization achieved seizure control, and the patient was discharged with neuro rehabilitation support. This case highlights the course of epilepsy in SWS, management challenges, and need for long-term multidisciplinary care.

Keywords: Sturge-Weber syndrome, Focal epilepsy, Breakthrough seizures, Pediatric neurology, Port-wine stain, Developmental delay

INTRODUCTION

Sturge–Weber syndrome (SWS) is a rare, sporadic neurocutaneous disorder resulting from somatic mosaic mutations in the GNAQ gene, causing aberrant vascular development in the skin, brain, and eyes (Figure 1).[1] It is characterized by facial port-wine stains, leptomeningeal angiomas, glaucoma, and progressive neurological dysfunction including seizures and developmental delay.[2] Epilepsy is the most frequent and disabling manifestation, occurring in approximately 75–90% of patients and often progressing to drug-resistant seizures, substantially impacting neurodevelopment.[3] Early diagnosis and coordinated multidisciplinary care are critical to improving long-term cognitive and functional outcomes.[4]

We present a case of a toddler with SWS who developed breakthrough focal seizures despite chronic antiepileptic therapy.

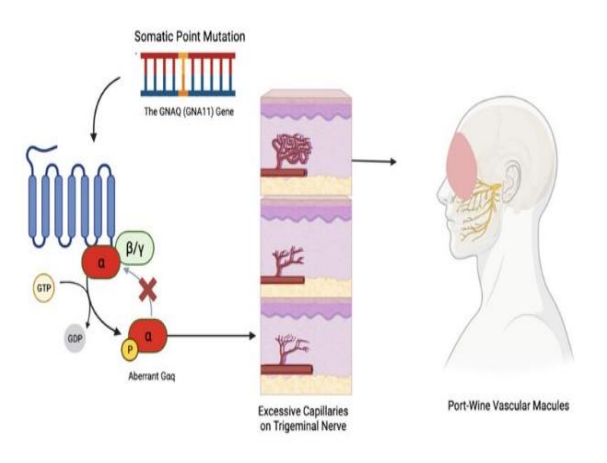

Figure 1: GNAQ gene mutation disrupts Gαq protein signalling, causing abnormal capillary proliferation.[1]

CASE REPORT

A 2-year-3-month-old boy, born to non-consanguineous parents and a known case of SWS with focal epilepsy, left-eye glaucoma, and global developmental delay, presented with three episodes of right-sided focal motor seizures with impaired awareness. Post-ictal drowsiness was noted, with no history of fever, missed medications, vomiting, or irritability. On examination, the child was drowsy with reduced spontaneous activity, generalized hypotonia, and paucity of movements on the right side, consistent with focal neurological deficit. A left hemifacial port-wine stain was present. Vital signs were stable and no signs of meningeal irritation or other systemic abnormalities were noted.

Seizure onset occurred at 5 months of age, with recurrent breakthrough episodes including status epilepticus at 10 months requiring PICU care. Antiseizure medications trialed included phenytoin (6 mg/kg/day), levetiracetam (50 mg/kg/day), valproate (20 mg/kg/day), clobazam, and phenobarbitone (20 mg/kg/day). Left-eye glaucoma was diagnosed during infancy and managed with topical dorzolamide-timolol drops.

Perinatal history revealed late-preterm delivery (36+4 weeks) via LSCS, with immediate cry at birth. He required NICU admission for two weeks due to respiratory distress and suspected neonatal sepsis. Neonatal hypocalcemic jitteriness and hyperbilirubinemia were treated appropriately. Initial echocardiography showed ASD/ patent foramen ovale (PFO), PDA, and severe pulmonary hypertension, which resolved by three months of age.

A latest computed tomography (CT) scan of the brain showed features consistent with SWS, including atrophy of the left cerebral hemisphere with prominent extra-axial CSF spaces (Figure 2), tram-track cortical calcifications (Figure 3) predominantly in the left frontal and parietal lobes, mild subcortical calcifications, and enlargement of the left choroid plexus. No evidence of intracranial hemorrhage, infarct, or midline shift was noted, and posterior fossa structures were normal.

Laboratory evaluation showed mild hypomagnesemia (Mg^{2+} 1.61 mg/dl), elevated blood ammonia (102 µg/dl), hypoalbuminemia (3.55 g/dl), and low total protein (5.29 g/dl). Hematology findings indicated mild anemia with Hgb 9.7 g/dl and reduced RBC count (3.43 million/µl), reflecting metabolic stress and nutritional compromise in the context of refractory seizures (Table 1).

Developmentally, the child had age-appropriate milestones until 6 months of age, followed by delays in gross motor, speech, and fine motor domains. On assessment, he was able to sit with support and stand with maximal assistance, vocalized 1–2 words, used an immature pincer grasp, and exhibited global developmental delay.

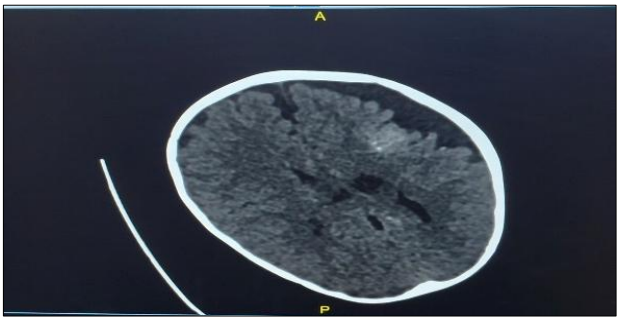

Figure 2: CT brain showing left cerebral atrophy.

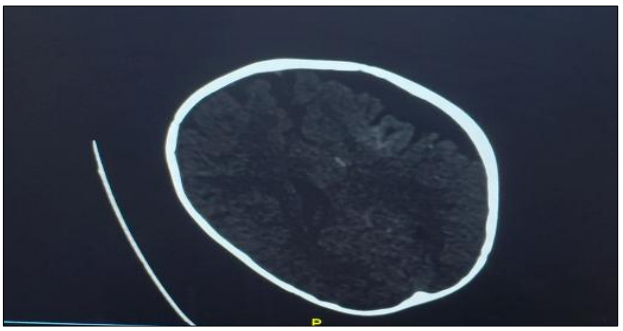

Figure 3: CT brain showing tram-track cortical calcifications.

Table 1: Significant hematological and bio chemical findings.

S. no.	Parameter	Child value	Reference range
1	Magnesium	1.61 mg/dl	1.7- 2.55 mg/dl
2	Blood ammonia	102. µg/dl	Upto 82 µg/dl
3	Albumin	3.55 g/dl	3.8-5.4 g/dl
4	Total protein	5.29 g/dl	6-8 g/dl
5	Total RBC	3.43 mil/microliter	3.6-5.2 mil/microliter
6	Hb	9.7 g/dl	10.8-12.8 g/dl

During hospitalization, antiepileptic therapy was optimized by increasing clobazam and adding phenobarbitone and phenytoin, resulting in good seizure control and improved sensorium.

The child was discharged with advice regarding strict medication adherence, regular pediatric neurology and ophthalmology follow-up, and continuation of neurodevelopmental rehabilitation (Table 1).

Nursing care plays a pivotal role in the management of children with SWS-related refractory epilepsy. In this case, nursing interventions focused on continuous seizure surveillance, maintenance of airway patency during ictal and post-ictal phases, prevention of injury, and strict adherence to antiepileptic medication schedules. Close monitoring for adverse drug reactions and neuro-vital changes was essential to identify early signs of deterioration. Developmental stimulation, physiotherapy coordination, and structured caregiver education on seizure first-aid, medication compliance, and home safety were integral components of care. Eye-care support, including correct instillation of glaucoma medications and monitoring for visual strain, enhanced ophthalmologic outcomes. Parental counseling and psychological support further strengthened family coping and long-term engagement with neuro rehabilitation and multidisciplinary follow-up.

Table 2: Medication on discharge.

S. no.	Name of the drug	Dosage and frequency
1.	Syp. Gardenal	2.5 ml - 0 - 5 ml
2.	Syp. Levipil	2 ml, BD
3.	T. Lacoset	25 mg, 1/2-0-1/2
4.	Syp. Tegrital	3.5 ml, BD
5.	T. Frisium	5 mg, 0-0-1
6.	Syp. Eptoin	3.5 ml-0-3.5 ml
7.	T. Ecosprin	15 mg, OD
8.	Syp. Calcimax P	0-2.5 ml-0
9.	Dorbet PF eyedrops	1 drops, BD

DISCUSSION

SWS results from post-zygotic GNAQ mutations, leading to abnormal vascular channel SWS results from post-zygotic GNAQ mutations that disrupt vascular maturation, leading to leptomeningeal angiomatosis, chronic hypoxia, and progressive cortical atrophy.[1,5] Neurological severity correlates with extent of cortical involvement and onset of seizures, which typically begin in infancy; early seizure onset (<1 year) is associated with poor developmental outcomes.[6] Our patient's seizures began at 5 months, aligning with literature describing early-onset epilepsy as a predictor of cognitive impairment and drug-resistance.

Neuroimaging classically demonstrates cortical atrophy, serpentine calcifications, and choroid plexus hypertrophy; CT is particularly sensitive in identifying calcifications, while magnetic resonance imaging (MRI) delineates leptomeningeal involvement.[4] Similar findings were seen in this child.

Drug-resistant epilepsy is reported in up to 60% of SWS cases and often necessitates polytherapy or advanced approaches including ketogenic diet, laser ablation, or hemispherectomy.[7,8] Recent studies emphasize early aggressive seizure control and early surgical evaluation in unilateral disease to preserve neurocognitive function.[9-11] Our patient required multiple antiepileptic medications due to breakthrough seizures, highlighting the challenge of seizure control in this population. Early rehabilitation, ophthalmic surveillance for glaucoma, and parental counseling form essential components of comprehensive management.[2,8]

Comprehensive pediatric neuro-nursing care plays a crucial role in optimizing outcomes in SWS-associated refractory epilepsy. Evidence underscores the importance of continuous seizure surveillance, timely airway support during ictal and post-ictal phases, monitoring for antiseizure drug adverse effects, and prevention of complications through early symptom recognition.[12]

Structured caregiver education significantly enhances home seizure management, improves medication adherence, and reduces emergency visits and readmissions in children with epilepsy.[13]

Family-centered nursing interventions and developmental-support programs further improve neurodevelopmental trajectories in children with chronic neurologic disorders.[14] In our case, proactive seizure monitoring, airway maintenance, medication compliance reinforcement, and parental coaching on seizure first-aid and glaucoma care aligned with evidence-based pediatric epilepsy nursing standards demonstrating the critical role of integrated neuro-nursing in multidisciplinary SWS management.[12,15]

Our case affirms the critical value of coordinated multidisciplinary care-including neurology, ophthalmology, developmental paediatrics, rehabilitation teams, and comprehensive neurodevelopmental nursing services in managing SWS. Timely seizure detection, proactive therapeutic optimization, and individualized rehabilitation planning are key determinants of favourable neurodevelopmental outcomes and long-term quality of life.

CONCLUSION

SWS is a complex neurocutaneous disorder in which early-onset, refractory epilepsy is a major determinant of developmental and functional outcomes. This case highlights the diagnostic features, therapeutic challenges, and importance of multidisciplinary management, including vigilant and evidence-based nursing care. Timely seizure management, consistent ophthalmic monitoring, and structured neuro-rehabilitation are critical. Empowering caregivers through education and long-term coordinated follow-up supports optimal disease control and improves neurodevelopmental prognosis in affected children.

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