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CASE REPORT

An Atypical Seizure Onset and Re-Emergence in a Refugee with an Undiagnosed Sturge-Weber Syndrome: A Case Report from a Limited Setting

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Video abstract

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Background: Sturge-Weber syndrome (SWS) is a complex rare genetic neuro-cutaneous disorder characterized by the presence of a port-wine stain, ophthalmic and intracranial angiomatosis leading to seizures, ocular, and oral abnormalities.

Case Presentation: We report a 39-year-old, non-diabetic, non-hypertensive female refugee who presented initially with heart failure due to anemia for which she received blood transfusions. Later on admission, she developed multiple focal to bilateral seizures, severe irritability, aphasia, and right-sided hemiplegia, leading to admission to the ICU. A repeat medical history and examination revealed a faint left-sided ophthalmic port-wine stain that was initially unnoticed and a remote history of unprovoked seizures 20 years ago. Imaging revealed parietal calcifications and confirmed the diagnosis of SWS. Thus, a multidisciplinary approach was taken to fully understand the patient's diagnosis and determine a treatment strategy, involving consultations with the neurology, ophthalmology, otolaryngology, and physiotherapy departments. Successful seizure control was achieved by administering IV phenytoin for 3 days and the up-titrating of oral carbamazepine to 1g daily through a nasogastric tube. Unfortunately, due to the unavailability of personnel or resources, other important assessments for patients with SWS, such as advanced neuroimaging, psychiatric, plastic and neuro-surgery evaluations, as well as dentistry reviews, could not be conducted.

Conclusion: This case highlights the rare occurrence of adult-onset seizures in an undiagnosed SWS and their re-emergence after almost two decades without anti-seizure medications. It also highlights the importance of a comprehensive history and clinical examination, as this patient's diagnosis of SWS could have been missed if she had not experienced seizures on admission. Our study also demonstrates the challenges associated with managing such a complex condition in settings with limited resources.

Keywords: genetic, angioma, seizure, glaucoma, port-wine stain

Introduction

Sturge-Weber syndrome (SWS), formerly known as leptomeningeal angiomatosis, is a rare, non-hereditary congenital neurocutaneous disorder caused by a mutation in the (GNAQ) gene. This gene is involved in angiogenesis and blood vessel function. This mutation can lead to different manifestations, including hamartomatous abnormalities affecting the central nervous system, skin, oral cavity, and eyes, usually in unilateral spread. SWS is primarily characterized by a facial capillary malformation known as nevus flammeus or port-wine stain (PWS), leptomeningeal angiomatosis, or glaucoma.[1] This condition is extremely rare, affecting approximately 1 in 20 to 50 thousand live births.[2] SWS most commonly presents with neurological manifestations, with seizures being the most predominant neurological feature in 80–90% of cases, intellectual disability in almost half of the patients, and focal neurological deficits (FNDs), such as hemiplegia and aphasia, which are usually transient.

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Case Presentation

A 39-year-old pregnant Ethiopian refugee (Gravida 3 Para 2+0) was referred to our internal medicine emergency department at Gadarif Teaching Hospital, Gadarif State, Sudan, with progressive generalized body swelling, shortness of breath, and fatigability for 6 weeks and an unremarkable systemic review. The patient was admitted to the critical cases admission site and her history, taken with the help of a language interpreter later on admission, revealed an initially undisclosed remote episode of three unprovoked generalized tonic-clonic seizures 21 years ago which was treated traditionally. However, there was no history of head trauma or a family history of seizures. General examination of the patient has revealed a left-sided PWS involving the ophthalmic division of the trigeminal nerve (Figures 1 and 2). However, this finding was initially missed on inspection but later confirmed by the patient's spouse to be present since birth. The patient was also severely pale and had bilateral pleural effusions, tender hepatomegaly, and bilateral lower limb swelling.

Initial investigations revealed a hemoglobin level of 8.84 g/dl. Thus, heart failure due to anemia was diagnosed, and the patient was treated accordingly. However, eight days after admission, the patient had developed approximately six episodes of focal to bilateral tonic-clonic seizures that responded well to phenytoin but rendered the patient in a deep comatose state with a GCS of three. She was subsequently admitted to the intensive care unit (ICU) and a nasogastric tube was inserted to aid with her nutrition and drug administration. Moreover, to prevent further seizures, she was commenced on carbamazepine tablets 200 mg administered through the nasogastric tube.

A non-contrast computed tomography (CT) scan of the brain was delayed to the day after stabilization because there was only a single CT scan machine in the state of Gadarif and it was operated intermittently due to the unavailability of

Figure 1 A photo of the patient's faint left ophthalmic port-wine stain.

Figure 2 A clear photo of the patient's left ophthalmic port-wine stain.

a sufficient number of radiology technicians. The CT scan revealed cortical calcifications in the posterior part of the left parietal area raising the differential diagnosis of SWS (Figure 3). Consequently, a repeat physical examination was performed, which showed the presence of a faint left-sided red to purple colored flat lesion on her forehead and eye. Upon questioning her spouse, this lesion was confirmed to be present and unchanged since birth, which helped us rule out the diagnosis of a salmon patch because this lesion did not fade, thus it was diagnosed as a PWS. Additionally, she had no features of megalencephaly capillary malformation syndrome which usually includes a large head or brain size, joint problems, and capillary malformations on the skin. Moreover, the patient had no limb hypertrophy, lymphatic drainage abnormalities, or bone or soft-tissue outgrowths suggestive of Klippel-Trenaunay-Weber syndrome. Thus, based on the CT scan findings and the typical presentation, we concluded the diagnosis of SWS.

Two days after ICU admission, the patient experienced a fluctuating change in consciousness and a new-onset contralateral hemiparesis (contralateral to the PWS), as well as aphasia, and emotional lability. Unfortunately, four days after admission to the ICU, the patient started to experience vaginal bleeding; hence, the obstetrics and gynecology department was consulted, and the patient was found to have a non-viable pregnancy (28 weeks + 1 d), and an uneventful induction was performed.

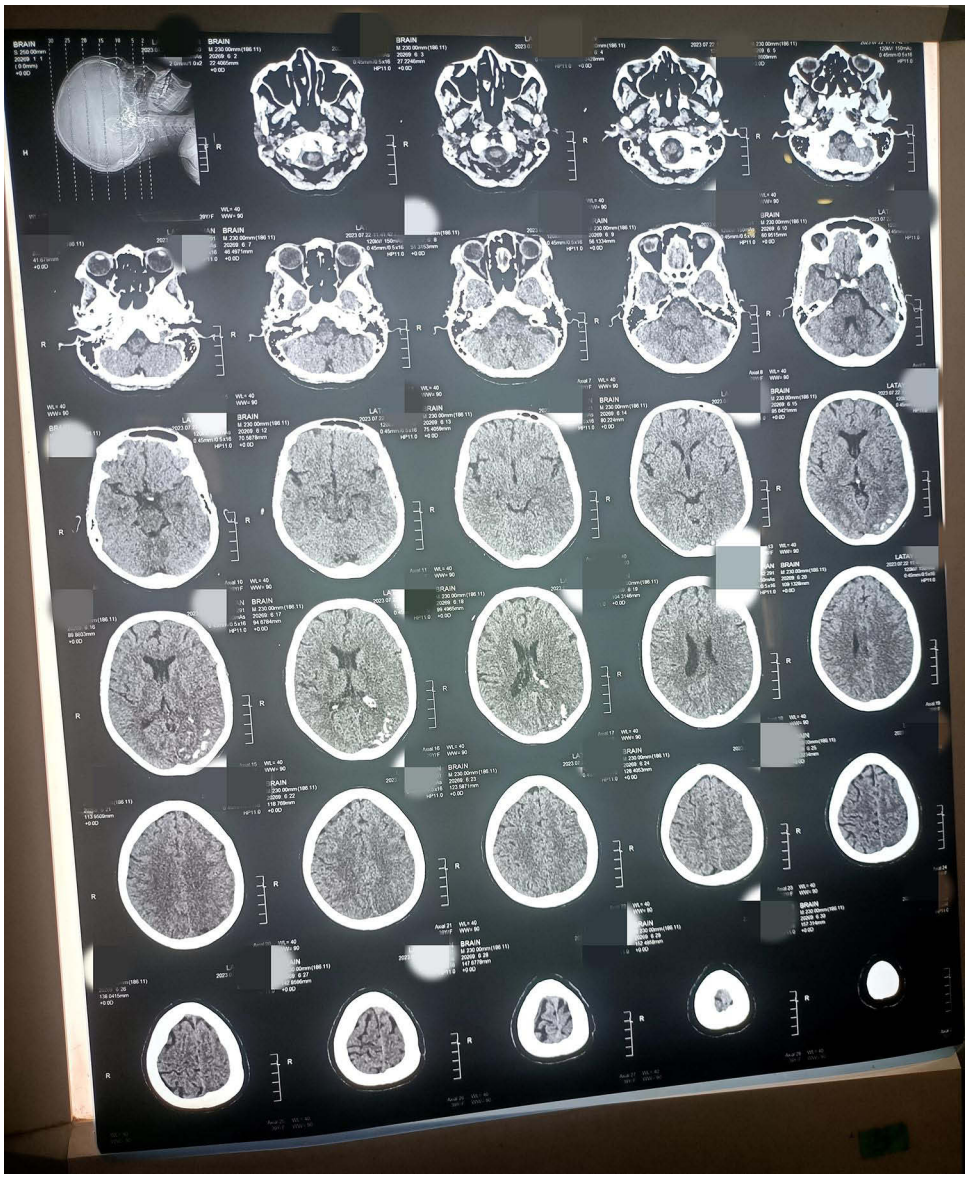

Figure 3 CT brain scan showing cortical calcifications in the left parietal area, 3rd and 4th rows.

During the same period, she became combative and uncooperative, which led to physical restraining. Three days later, the patient developed new focal to bilateral seizures that started as facial myoclonic episodes, and after consultation with the neurology department, the patient's dose of carbamazepine was increased to 1000 mg daily, which helped in preventing seizure recurrence. It is worth mentioning that in her second trimester, and before receiving the CT scan result, a diagnosis of eclampsia was explored, but her consistent blood pressure readings with an upper normal systolic blood pressure of 139 on most days and a diastolic pressure with a maximum of 97 mmHg and a low range of proteinuria, as well as the absence of systemic features such as renal or liver impairment, excluded this diagnosis. After reaching the diagnosis of SWS, a multidisciplinary team (MDT) approach was put into play; hence, an ophthalmology consultation was performed to exclude glaucoma. However, the patient was very irritable and difficult to perform a fundoscopy examination. Moreover, the patient was transferred to the internal medicine wards after stabilization.

Discussion

Classification

According to the Roach Classification, SWS can be classified into three distinct types, depending on the presence or absence of facial and/or central nervous system angioma and glaucoma.[3] Type 1 SWS includes the presence of facial PWS, vascular malformation of the brain (intracranial leptomeningeal angioma), and possible glaucoma. While type 2 SWS is thought to be the most common subtype, it consists of facial angioma and the possibility of glaucoma, but an absence of brain involvement. Type 3 SWS is described as the occurrence of leptomeningeal angioma without PWS and classically without the development of glaucoma or any other eye abnormalities. Thus, our patient most likely has type 1 SWS.

Clinical Presentation

Clinical manifestations of SWS syndrome are very diverse, from simply having only a PWS to a full-blown picture of neuro-oculo-psychological presentations such as seizures, migraine-like headaches, intellectual disability, and stroke-like events. Additionally, patients may have mood swings and glaucoma.[4] Although, in SWS, seizures are one of the earliest manifestations and usually occur by the first year of life in almost 75 to 90%[5] of patients and are usually difficult to control if not treated early on, our patient's case is interesting and unique as she had her first unprovoked seizure episodes around the age of 18 to 20 years then was seizure free without treatment till presentation to our hospital at 39 years of age, moreover, her seizures were relatively easy to control. Nevertheless, we found an even stranger presentation of a case that reported the onset of tonic-clonic seizures in the sixth decade of life in a patient with a type 3 SWS.[6] Although port-wine stain is usually the leading evidence for the diagnosis, it was initially unnoticed on general examination of our patient, this could be due to the complexion of the patient or more possibly the rarity of the condition. However, it was detected later on after repeating a careful history and examination following the development of seizures during admission. It is worth mentioning that PWS affecting the complete ophthalmic distribution of the trigeminal nerve (V1) was highly predictive of ocular and/or neurological affection (78%), whereas partial affection of (V1) was found to decrease the risk to 26%.[7] Moreover, although there are many documented oral presentations of SWS, including histological and morphological changes, the most common abnormality of which is gingival hyperplasia affecting the maxilla, lips, palate, and tongue on the ipsilateral side.[8,9] However, due to the irritability, combativeness, and emotional lability of the patient, these oral presentations were not examined and a proper ophthalmologic examination could not be conducted, this was exacerbated by the fact that the ophthalmology and dentistry departments were located in separate hospitals. In conclusion, our patient presented with a myriad of typical features of SWS, including the PWS, focal-to-bilateral seizures, hemiparesis, and aphasia.

Radiological Findings and Diagnosis

After careful history taking and physical examination, when suspicion of SWS is raised, neuroimaging is performed to detect any intracranial lesions and delineate the severity of patients' conditions. Magnetic resonance imaging (MRI) was found to be superior to CT in detecting intracranial malformations.[10] Moreover, A CT or MR angiography is required to assess the presence or absence of intracranial angiomas. However, in the limited setting that we had and because of the

absence of more advanced technology, we were only able to organize a non-contrast CT brain that revealed intracranial calcifications in the left posterior parietal area. The unavailability of well-equipped healthcare facilities sheds light on the incapacity to outline the full extent of the disease for patients with SWS, which may be more troublesome for a refugee, such as our patient, and significantly affect the prognosis and endanger the lives of such patients.

Management

The Management and prognosis of patients with SWS are highly dependent on their signs and symptoms and are usually performed using an MDT approach. Management options may include anti-seizure medications, ophthalmologic therapies such as eye drops, or eye surgery for glaucoma, a condition that can be seen in up to 70% of cases with SWS.[11] Additionally, some patients may be offered laser therapy or plastic surgery for PWS, dental care, and rehabilitation in terms of delineating the patients' dental, gingival, and oral hygiene conditions, as well as physical and speech therapy for the focal neurological deficits that may emerge, such as hemiplegia or aphasia.

Management of this specific case was very challenging because of the late incidental presentation, the limited setting in which the patient was treated, the unattainability of important diagnostic tests, and the lack of in-hospital departments that should have been included in the MDT approach, such as dentistry, plastic and neuro-surgery, and psychiatry. Hence, the full picture of our patient's condition was not understood. Follow-up of patients with SWS is paramount to a good prognosis. Starting with good counseling and explanation of the nature and genetics of the disease, the fluctuating course of symptomatology such as focal neurological deficits, and the importance of regular follow-up and visits to the relevant clinical departments are vital to better patient care. Refugee conditions, such as in this particular case, make it very challenging to provide the best standard of care to patients. However, after careful counseling of the patient's spouse, although prematurely, our patient was discharged on carbamazepine tablets 200 mg BD, and a request form was written to the referring refugee organization explaining the need of our patient for more advanced neuro-imaging, a good ophthalmic and dentist evaluation, a visit to a dermatologist, and frequent follow-up appointments with a neurologist and a physiotherapist. Our patient was seen three weeks later for follow-up, during which, she was seizure-free, and her hemiplegia and aphasia have started to improve as now she was able to walk with assistance and able to produce speech.

Conclusion

Sturge-weber syndrome is an extremely rare and complex condition that may present with various manifestations and complications. We believe that this case is unique in terms of the atypical seizure onset in our patient at 18 years, who also remained seizure-free for almost 20 years after her initial seizure presentation despite not receiving any anti-seizure medications. Further studies may be required to investigate whether acute illnesses may be a factor in seizure re-emergence in patients with SWS. This study is also a reminder that a comprehensive history and clinical examination are essential and remain vital to the management of all patients and that with the aid of good clinical sense and reasoning, they can lead to appropriate diagnoses and effective management. Furthermore, in-depth knowledge of SWS and its different presentations and complications is paramount to understanding the full spectrum of this condition and to be able to deliver the best care possible to such patients. We also highlight the impact of the limited resources in managing such patients in terms of the lack of advanced neuro-imaging techniques and different components of a multidisciplinary team stressing the importance of improving access to a better healthcare system.

Ethical Approval

This was a single case report and institutional board approval was not required to publish the patient's details. Because our patient was aphasic and in profound emotional lability, we were not able to counsel her regarding her condition or obtain ethical approval from her. Moreover, she was physically combative, she also had a single caregiver who was her spouse. Unfortunately, and against medical advice, the patient's spouse has asked repeatedly for a premature discharge, hence, the ethical approval for publication of the patient's current condition, past medical history, and the images and figures included within the manuscript was obtained from her spouse on discharge for clinical case presentation and report using an adapted BMJ case report consent form signed by her spouse.[12] This form will be possible to view on reasonable request.

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Disclosure

All authors report no conflicts of interest in this work.

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