

Sturge-Weber Syndrome with Bipolar Presentation: A Case Report

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Abstract

Introduction: Sturge-Weber Syndrome (SWS) is a rare neurocutaneous syndrome that is manifested by overt neurological and covert psychiatric features. Although the syndrome is known to be neurocutaneous, multiple organs and systems are involved.

Case Presentation: A 45-year-old male, with type I SWS was admitted to the psychiatric ward with manic-like symptoms. The case had a history of repeated psychiatric admissions due to seizures, mood and behavioral disorders. Upon admission, he had elevated mood, talkativeness, increased libido along with ictal aggression with psychic aura. The symptoms remitted with anticonvulsants. Other required diagnostic and treatment workups were also conducted.

Conclusions: A holistic approach should be adopted to manage these patients. Left brain involvement, the associated stigma, seizure disorder, adverse effects of antiepileptic medications along with coping with a chronic condition might have been the underlying mechanisms for mood and behavioral changes in this patient.

Keywords: Bipolar Disorder, Case Report, Sturge-Weber Syndrome

1. Introduction

Sturge-Weber Syndrome (SWS) is a rare neurocutaneous syndrome that is manifested by overt neurological and covert psychiatric features. It usually occurs sporadically with equal prevalence in both sexes (1, 2).

In neonates, the main presentation is usually the cutaneous angiomas, frequently seen in the distribution of the ophthalmic branch of the trigeminal nerve and is called the Port-Wine stain (3, 4). The Port-Wine stain is seen in 98% of the patients, but not all patients with Port-Wine stain have SWS (5, 6).

In early childhood, the disease is associated with delayed milestones and seizures (6). Seizures occur in 83% of the patients. Seizures in these patients commonly have focal onset but can become generalized (7). When the seizure onset is before the age of two, cognitive impairment and learning disability are more frequently seen (5).

Recurrent thrombosis and stroke-like events are other concerns in these patients. Antiplatelet medications could reduce thrombosis and promote perfusion (8). The frequency of cognitive decline, hemiparesis, seizure and other neuropsychiatric sequels of stroke-like events may decrease by provision of antiplatelet medications.

Screening for endocrinological abnormalities should be conducted during childhood. These abnormalities include thyroid dysfunction, growth hormone deficiency and probably impaired glucose metabolism (1).

Glaucoma is another common manifestation (30% - 60%), which is usually open-angled and ipsilateral to the Port-Wine stain and can result in progressive vision loss (6).

In paraclinical evaluation and Brain MRI, angiomas are seen ipsilateral to the cutaneous lesion. In brain CT, calcification is seen in parietal and occipital lobe (3).

During adulthood, psychiatric and behavioral manifestations, including mood, anxiety, adjustment and disruptive behavior disorders (DBD), cognitive deficits, sleep problems and substance-related disorders, become more prominent (1, 9).

Usually the patients are managed by a neurologist for their seizures, but psychiatric features are commonly neglected. Although SWS is known as a neurocutaneous syndrome, multiple systems and organs are involved. The aim of this case report was to emphasize the importance of a holistic diagnostic and therapeutic approach in such cases. Although behavioral symptoms are common in this syndrome, the manifestations of this case resembled a bipolar disorder, and the underlying predispositions could have been missed.

2. Case Presentation

A 45-year-old male was admitted to the psychiatric ward due to aggression. He was never married, had only received home-based education and used to have a part-time

simple job. From a year ago, the patient had developed elevated mood, talkativeness, verbal and physical aggression, increased libido and disinhibited behaviors. Some of the aggressive spells were non-purposeful and the patient had no recollection of the events.

By reviewing the past medical and psychiatric history of the patient, we found that he had experienced asphyxia at birth, following a complicated natural vaginal delivery. He had a congenital Port-Wine stain on the left side of his face. At the age of five months, the patient experienced onset of seizures during febrile episodes. The seizures were characterized by lip smacking and eye blinking and developed into generalized tonic clonic seizures (GTC). In addition, the patient's milestones were grossly delayed. He was unable to pass the first grade in primary school for three consecutive years and was not able to attend school. The patient had behavioral problems, verbal aggression and oppositional behavior during childhood. At the age of 10, he was hospitalized due to seizures and behavioral problems. The seizures were characterized by ictal fear, upward gaze and falling to the right side. In postictal phase, he had no recollection of the events and experienced headache in the left side.

When the patient was 25, he had a suicidal attempt by consuming too many pills. The act of suicide seemed to be rather impulsive, following a relational conflict with the family with no significant mood changes at that time.

At the age of 36, he was admitted to a psychiatric ward for the second time again due to his aggressions. At that time, he was seizure free and was discharged with the diagnosis of "personality changes due to a general medical condition".

During his third admission at the age of 41, he began to experience stereotypic visual hallucinations as aura by seeing a "lion", followed by a secondary GTC. The patient's seizures were controlled by carbamazepine 1200 mg and phenytoin 200 mg. In addition, metformin 1000 mg daily was initiated for his newly diagnosed diabetes mellitus. No major psychiatric or medical conditions were detected in the family history.

Upon admission, the patient's mood was elevated in mental status examination (MSE). He showed signs of irritability and impulsivity during the interview. Speech had increased volume. In cognitive evaluation, he scored 24/30 in the mini mental status examination (MMSE). In montreal cognitive assessment (MoCA), he scored 11/30 and had deficits in recent memory, attention, abstract thinking, visuospatial and executive functioning. In Wechsler Intelligent Scale, he scored 69, 71 and 68 in verbal, performance and overall IQ, respectively.

Brain CT revealed calcification in the left occipital lobe. In the brain MRI, the left choroid plexus was enlarged com-

pared to the right side and white matter changes, and atrophy were seen in the left occipital lobe with no obvious enhancement. Subtle abnormal signal intensity was seen in the left hippocampus when compared with the opposite side. Similar to other studies, MRI scoring system was used for this patient (9, 10). Left side MRI score was 6/16 and total MRI score was 10/32. The patient's score in SWS neurological assessment is summarized in Table 1.

Table 1. SWS Neurological Assessment

SWS Neurologic Assessment	Patient's Score	Description
Visual field cut	1/2	Partial homonymous hemianopia
Seizure frequency	2/4	Breakthrough
Hemiparesis	0/4	No hemiparesis
Cognitive functioning	3/5	No work, academic functioning
Total score	6/15	

Abbreviation: SWS, Sturge-Weber Syndrome

A lower score correlates to better overall neurologic function and lower disease severity. Cognitive function score correlates to total MRI score, left occipital and left frontal subscore (11).

EEG study revealed background slowing in theta range in the left side, but unexpectedly epileptiform discharges were seen in both sides. As the patient experienced recurrent anger spells without recollection of the events in the presence of EEG abnormalities, we considered complex partial seizure (CPS) with psychic aura. Therefore, carbamazepine was raised to 1400 mg per day, which resulted in mood stabilization and the remission of aggressive behavior.

In dermatological evaluation, treatment with pulsed dye laser (PDL) was recommended. In ophthalmological examination, right hemianopia was seen in perimetry while the intraocular pressure was found to be within normal range. Required workup was also provided for the treatment of his diabetes mellitus. Abdominal and pelvic ultrasound study did not reveal any pathological finding.

3. Discussion

SWS is commonly manifested by neurological features such as seizures and developmental delay (1). Psychiatric features of this syndrome are often overshadowed by other dermatological and neurological signs. In this case, the chief complaints and the reason for referral were psychiatric and behavioral symptoms, which led to the diagnosis of SWS.

Our case had a Port-Wine stain in the distribution of the ophthalmic branch of left trigeminal nerve, which spreads to the left side of the forehead and left upper eyelid (maxillary branch). Therefore, according to Roach classification, he had type I SWS (2). The prognosis is worse in patients with Port-Wine stain, stretching from the midline of the forehead to the line joining the outer canthus of the eye to top of the ear (12).

Characteristically, leptomeningeal angiomas occur as a unilateral lesion affecting posterior temporal, parietal and occipital areas (1). Venous angiomas show abnormal blood flow pattern and results in gliosis, atrophy and progressive calcification that produce the characteristic tram track line in gyriform pattern (13). These lesions may cause seizure or hemiparesis. Our patient had experienced seizures from birth up to three years ago. In recent years, seizure pattern had changed to a stereotypic simple visual hallucination. Generalized seizures occurred monthly and responded well to carbamazepine and phenytoin. Thus, the patient's aggression was considered partly as an ictal phenomenon with psychic aura. In neuroimaging, hyperintensities were seen in the left occipital region, which could correlate with atypical seizure type: Stereotypic visual hallucination.

The observed learning difficulties and intellectual disability in this patient might be due to his early onset seizures, adverse effects of antiepileptic medications and left brain involvement (1). Other studies and reports have also reported similar findings. Early onset of seizures and left brain involvement are correlated with poor cognitive outcomes. Mental retardation presents in 50% - 60% of these patients (14-16). Attention deficits have been reported in 50% of these patients (1).

Mood disorders, disruptive behaviors and suicidality are common in this syndrome (1). Our patient also reported a history of depression and suicidal behavior. Left brain involvement, his birthmark and associated stigma, seizure disorder, adverse effects of antiepileptic medications along with coping with a chronic condition have been proposed as the underlying mechanism for this observation.

Disruptive behavior is another common psychiatric manifestation in SWS (1, 17). Some studies have highlighted verbal aggressions and legal problems. Aggression is more commonly seen in SWS patients with mental retardation. The proposed mechanisms for this association quite resemble those for mood and cognitive problems. However, other medical comorbidities should be considered. When the patient has transient non-purposeful aggressions, he/she should be screened for atypical partial seizures. Glaucomas, which sometimes can be quite painful, are considered to be another source of distress and

disruptive behaviors in these patients (18). Therefore, regular ophthalmic examination should be considered.

In conclusion, clinical presentations of SWS can be both psychiatric and neurologic. Clinicians should adopt a holistic diagnostic and treatment approach. Other work-ups, including dermatologic, ophthalmologic and consultation with an internist should also be conducted.

Footnotes

Authors' Contribution: Zahra Mirsepassi, Fatehemeh Mohammadian and Behrang Shadloo did the literature review and drafted the manuscript. Zahra Mirsepassi, Elham Hakki and Behrang Shadloo collected the clinical data and presented the case. All authors read and approved the final manuscript.

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