Vol 4 | Issue 3 | July - September | 2024

CASE REPORT - PUBLIC HEALTH

MULTI DISCIPLINARY APPROACH IN STURGE-WEBER SYNDROME: A CASE REPORT

Deleepa R [1]

(1) Assistant Surgeon - Dental, Upgraded Government Primary Health centre, Adhanakottai, Pudukkottai HUD

ABSTRACT

INTRODUCTION : Sturge-Weber Syndrome (SWS) is a rare congenital neurocutaneous disorder characterized by facial capillary malformations and associated neurological complications. This case report details a 24-year-old female with Sturge-Weber syndrome (SWS) who sought cosmetic surgery to address aesthetic concerns stemming from facial disfigurement, including facial palsy and a port-wine stain. Comprehensive evaluation revealed a rash consistent with SWS, alongside a history of seizures. The report emphasizes the need for a multidisciplinary approach to manage both cosmetic and medical challenges while highlighting the importance of understanding the implications of dental procedures in such patients.

INTRODUCTION

Sturge-Weber syndrome (SWS) is a congenital neurocutaneous syndrome defined by the association of a facial capillary malformation in the trigeminal nerve's ophthalmic distribution with vascular malformation of the brain, eye and ipsilateral vascular glaucoma1. The Sturge-Weber syndrome and port-wine stains are thought to be caused by somatic mosaic mutations that interfere with vascular development2. SWS is a rare condition with an incidence of 1 case per 50,000 populations 2. The presentation of SWS often includes distinctive facial features that may significantly impact an individual's self-esteem and quality of life. This case report details the clinical findings and considerations in managing a 24-year-old female patient with SWS who sought cosmetic intervention to improve her facial aesthetics.

CASE REPORT

A 24-year-old female presented to our Dental OPD with several notable concerns. She exhibited facial palsy on the right side, swollen lips, and a reddish stain that extended from her forehead to her chin on the same side, reaching halfway across her hard palate without crossing the midline. This patient sought to enhance her appearance through cosmetic surgery. Her medical history revealed that she had a rash on the right side of her face since birth, which had progressively enlarged and darkened over the years. Additionally, she had a history of seizure episodes and was currently on anti-epileptic medication.

Upon physical examination, the right side of her face displayed a diffuse, flat reddish-purple rash affecting the areas supplied by the three branches of the trigeminal nerve. The rash extended from the midline of her forehead, up to the hairline, down to the lower border of her mandible, laterally to the external ear, and medially to the midline of the chin. The discoloured patch had an irregular shape with well-defined margins. The upper lip was notably swollen and everted, showing blanching when pressure was applied. When palpated, the patch felt flat, smooth, non-tender, and warm to the touch, with no visible pulsations detected. Patient visited our dental clinic to improve her facial aesthetics through facio-maxillary and cosmetic surgery. But as any dental procedures in these patients poses challenges due to potential risk of haemorrhage, she was explained about the complications and advised only about maintenance of good oral hygiene.

DISCUSSION

SWS is characterized by leptomeningeal angiomatosis and port wine stain of the face in the region of the trigeminal nerve's ophthalmic distribution4. Other neurological symptoms include seizures, hemiparesis, recurrent headaches, stroke-like episodes, psychomotor retardation, and mental retardation.[5] SWS may lead to cognitive impairments, visual field deficits, stroke-like episodes, endocrine problems and learning difficulties. Cutaneous manifestation (port-wine stain) in the distribution of ophthalmic and maxillary divisions of the trigeminal nerve

Please Scan this QR Code to
View this Article Online
Article ID: 2024:04:03:12
Corresponding Author: Deleepa R
e-mail : deleepa@gmail.com

is common in SWS. In 90% of patients with SWS, the first manifestation is seizure (focal or generalized) which occurs in the first year of life.

Although the aetiology is not well established, a previous study conducted has shown that a somatic activating mutation in the GNAQ gene is responsible for SWS.[3] The persistence of immature sinusoidal vascular channels and underdeveloped superficial venous drainage with compensatory dilatation of venules leading to shunting of blood to deeper veins, stasis and ischemia ultimately resulting in seizures, transient hemiparesis and progressive deposition of calcium salts is the postulated pathogenesis for SWS.[4]

Radiological imaging with the findings of leptomeningeal vascular malformation on contrast-enhanced T1-weighted magnetic resonance imaging (MRI), and cortical and subcortical calcification on head computed tomography (CT) is the basis of diagnosis.6 EEG, magnetic resonance spectroscopy and fluorodeoxyglucose-positron emission tomography (FDG-PET) may also help in the evaluation of patients but are not routinely used and are preferred. MRI is the most preferred modality for diagnosis in patients aged above one. CT scan detects calcification, gyriform calcification being the most common feature and described as a "tram-track sign". The use of ionizing radiation in CT scans limits its use.[6]

The treatment modality is generally supportive and symptomatic management using antiepileptics and aspirin is done.[7] Surgical procedures, which include hemispherectomy, corpus callosotomy, vagal nerve stimulation, focal resection of seizure focus (contraindicated in patients with bilateral involvement), are reserved for people not responsive to medical therapy and patients with glaucoma, refractory seizures and scoliosis. However, most patients achieve seizure control with medications.

A major component of Sturge Weber Syndrome is the significant facial disfigurement caused by soft tissue and bony overgrowth secondary to the associated vascular malformation. The surgical treatment of this deformity, however, has received little coverage in the literature8. Even with full patient cooperation, appropriate surgical intervention by reconstructive surgeons to restore normal anatomy and improve quality of life is extremely challenging and requires multi-staged procedures9. Patients with Sturge Weber Syndrome may also suffer from significant seizures due to intracranial involvement as well as psychosocial distress related to the disfigurement and resultant social stigmatization.[10]

This case provides valuable insights into the clinical presentation and diagnosis, of SWS in a young patient. We learned that any dental procedures might lead to life threatening complications. A multidisciplinary approach might be the solution to improve the facial aesthetics.

A comprehensive understanding of the latest treatment methods and a thorough knowledge of this syndrome are crucial for effectively managing this condition. The clinical observations in this case enhance our understanding of SWS and emphasize the importance of being proactive in early intervention for such cases.

Informed written consent was obtained from the patient regarding, protected health information for scientific presentation and publication purpose. It was obtained after explaining what information will be used, how it will be used, and where it will be published and patient was aware that their anonymity cannot be guaranteed.

LIMITATIONS

Given that this is a single case report, there is a potential for anecdotal fallacy, making it challenging to generalize the findings. Additionally, the patient did not return for follow-up, so we could not obtain further information regarding her progress or the outcomes of her treatment.

CONCLUSION

This case highlights the complexities involved in diagnosing and managing Sturge-Weber Disease, particularly regarding aesthetic concerns and the challenges posed by dental treatment. A thorough understanding of the condition and its implications for dental procedures is crucial for effective patient management.

REFERENCES

1. Dulac O, Lassonde M, Sarnat HB. Handbook of clinical neurology [Internet]. Amsterdam: Elsevier; 2013. [Aug 22; 2023]. pp. 315–21.Chapter 37, Sturge-Weber syndrome; pp. 31521.https://www.sciencedirect.com/science/article/pii/B9780444528919000373 Available from: [CrossRef] [Google Scholar]

2. Tonn JC, Grossman SA, Rutka JT, Westphal M. Neuro-Oncology of CNS tumors [Internet]. Berlin: Springer-Verlag; 2006. [Aug 22; 2023]. Neurocutaneous syndromes; pp. 327–43.http://link.springer.com/10.1007/3-540-31260-9_20 Available from: [Google Scholar]

3. Shirley MD, Tang H, Gallione CJ, Baugher JD, Frelin LP, Cohen B, et al. Sturge-Weber syndrome and port-wine stains caused by somatic mutation in GNAQ. N Engl J Med. 2013 May 23;368(21):1971–9. doi: 10.1056/NEJMoa1213507. [PMC free article] [PubMed] [CrossRef] [Google Scholar]

4. Bachur CD, Comi AM. Sturge-Weber syndrome. Curr Treat Options Neurol. 2013 Oct 1;15(5):607–17. doi: 10.1007/s11940-013-0253-6. [PMC free article] [PubMed] [CrossRef] [Google Scholar]

5. Marana Perez AI. Ruiz-Falco Rojas ML, Puertas Martin V, Dominguez Carral J, Carreras Saez I, Duat Rodríguez A, et al. Analysis of Sturge-Weber syndrome: a retrospective study of multiple associated variables. Neurol Engl Ed. 2017 Jul 1;32(6):363–70. doi: 10.1016/j.nrleng.2015.12.006. [PubMed] [CrossRef] [Google Scholar]

6. Comi AM. Presentation, diagnosis, pathophysiology and treatment of the neurologic features of Sturge-Weber Syndrome. The neurologist. 2011 Jul;17(4):179–84. doi: 10.1097/NRL.0b013e318220c5b6. [PMC free article] [PubMed] [CrossRef] [Google Scholar]

7. Comi AM. Update on Sturge-Weber syndrome: diagnosis, treatment, quantitative measures, and controversies. Lymphat Res Biol. 2007;5(4):257–64. doi: 10.1089/lrb.2007.1016. [PubMed] [CrossRef] [Google Scholar]

8. Surgical treatment of head and neck port-wine stains by means of a staged zonal approach. Cerrati EW, O TM, Binetter D, Chung H, Waner M. Plast Reconstr Surg. 2014;134:1003–1012. [PubMed] [Google Scholar]

9. Vascular malformation of the face: a rare case with follow-up of 18 years. Almeida de Arruda JA, Andrade ES, Andrade RA, Pereira JRD, Callou G. Acta Stomatol Croat. 2017;51:332–335. [PMC free article] [PubMed] [Google Scholar]

10. The removal of angiomas or port-wine stains from the face; associated ptosis of the lid and its correction. Rowland AL. Am J Surg. 1956;92:849–851. [PubMed] [Google Scholar]