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Case report

Art of port wine birthmark: a case report on type 2 Sturge–Weber Syndrome

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Abstract

Sturge–Weber syndrome is a phakomatosis group of neurocutaneous disorders. They manifest in the dermal, neural, ocular, and oral regions because of a mutation in the guanine nucleotide-binding protein G(q) subunit alpha gene. These lesions are often unilateral, following the course of the trigeminal nerve. Here, we present a case of a 54-year-old woman with a history of glaucoma with a unilateral reddish–purple discoloration of the palate and a port-wine stain limited to the left side of her face. The clinical course of Sturge–Weber syndrome is complex and requires a multidisciplinary approach. In type 2 cases of Sturge–Weber syndrome, patients present with facial angiomas and glaucoma, but without neurological involvement. There is an increased risk of hemorrhage associated with Sturge–Weber syndrome, which makes it essential to carefully plan dental procedures to minimize any potential pre and postoperative bleeding risks. This case report aims to highlight the importance of recognizing similar patterns to enable prompt diagnosis.

Keywords: Angioma, capillary malformations, glaucoma, palatal, port wine stain, vascular lesions.

Sturge–Weber syndrome is a rare congenital noninherited disorder that occurs in 1: 50,000 live births. They are also referred to as meningofacial angiomatosis, encephalotrigeminal angiomatosis, and encephalotrigeminal angiomyomatosis.[1] In addition to being a phakomatosis disorder, it is etiologically linked to a mutation in the guanine nucleotide-binding protein G(q) subunit alpha (*GNAQ*) gene on chromosome 9q21. This results in a triad of features involving ocular, cutaneous, and neurological symptoms, including increased intraocular pressure, facial port-wine stain, and leptomeningeal angiomatosis.[2] Schirmer was the first to give a precise description of this syndrome in 1869, and Sturge elaborated on this in 1879 with the inclusion of ophthalmic and neurological manifestations. Insight into the radiographical interpretation was highlighted by Weber in 1929.[3, 4]

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As early as 1992, Steve Roach proposed a classification based on the manifestations. Type 1 is the most common, involving facial and leptomeningeal angiomas, which may include glaucoma. Type 2 does not have intracranial involvement, with only facial port wine stains and glaucoma. Type 3 cases only suffer from intracranial involvement with no facial angiomas.[5] These lesions are usually unilateral and often follow the course of the trigeminal nerve. In addition, the oral cavity involves the presence of purplish-red macules that affect the gingiva, buccal mucosa, palate, and floor of the mouth. Other reported findings include angiomatosis that causes pyogenic granuloma, gingival hyperplasia due to the intake of anticonvulsant drugs, and malocclusion.[6] It is necessary to plan dental procedures with caution, as this disorder can manifest in different ways. In light of the above, here we report on a case of type II Sturge–Weber syndrome that presented with oral, ocular, and facial manifestations.

Case report

A 54-year-old female reported to the dental outpatient department with a complaint of pain in her upper left tooth that had persisted for the past week. Her medical history revealed a history of glaucoma involving the left eye, and she was under ophthalmic management. The family history was noncontributory. However, extraoral examination revealed unilateral deep purplish-red pigmentations localized to the left side of the face, ear, temple, and neck extending to the upper torso without crossing the midline **(Figure 1).** A history of pigmentations was noted since childhood, with a remarkable transition in color intensity from pink to purple as her age progressed. There was no significant history of bleeding or paresthesia in the pigmented regions.

The intraoral examination of the palate revealed a reddish–purple discoloration with the lesion extending anteriorly from an imaginary line connecting the 26 and 16 of the hard palate to posteriorly involving the soft palate region, but without crossing the midline **(Figure 2).** Other intraoral areas appeared to be normal. Upon palpation, the lesions were nontender, soft, and exhibited blanching; thus, the diascopy test was positive with no signs of pulsations. Dental findings included multiple missing teeth, dental caries with grossly destroyed 23, and gingival recession. Based on the clinical findings and medical history, a diagnosis of Sturge–Weber syndrome was considered. Further, the patient was advised to undergo posteroanterior skull and lateral skull radiographs to investigate the presence of any tram track calcifications, but these did not reveal any positive radiographic findings **(Figure 3).** Furthermore, the case was discussed with a neurologist for an opinion before arriving at a final diagnosis of type 2 Sturge–Weber syndrome. Based on the radiographic findings of the posteroanterior and lateral views, which revealed no evidence of calcifications, the patient was not advised to undergo computed tomography or magnetic resonance imaging at present. However, the patient was kept under observation with regular follow-ups, and advanced imaging will be planned if symptomatic. The patient was educated about the diagnosis and possible precautions to be taken before proceeding with the dental procedures, including oral prophylaxis, extractions, and prosthesis work. The patient was kept under review and is scheduled for regular follow-ups.

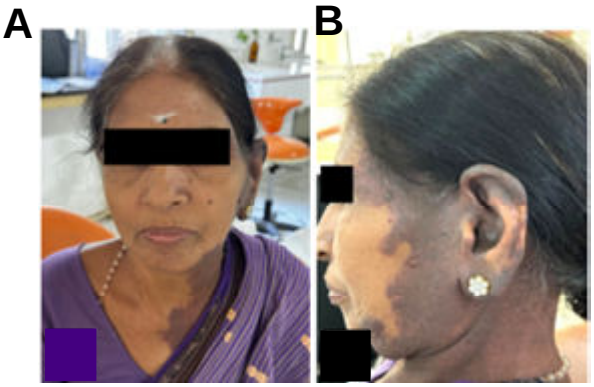

Figure 1. Unilateral port wine stain **(A)** antero-posterior view **(B)** lateral view.

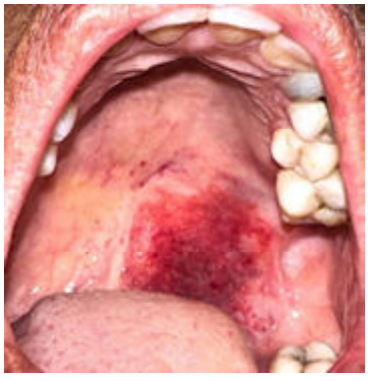

Figure 2. Unilateral presentation of intraoral palatal angioma.

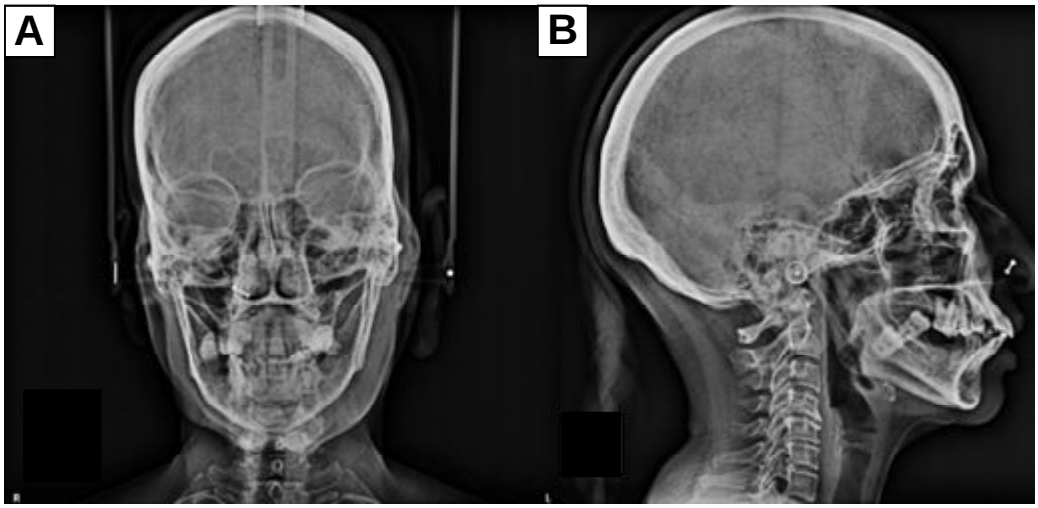

Figure 3. Radiography of skull in this patient **(A)** antero-posterior view **(B)** lateral view.

Discussion

Sturge–Weber syndrome is a rare neurocutaneous disorder that presents with dermatological, neurological, oral, and ocular manifestations. It is considered the third most common neurocutaneous syndrome after neurofibromatosis and tuberous sclerosis.[7] Abnormal signal pathways in GNAQ or G protein subunit alpha 11 variations lead to high endothelial activity during the embryonic stage. This abnormal signaling is thought to impair endothelial differentiation and induce capillary overgrowth and progressive dilation of immature venule-like vasculature in the arteries.[8] This syndrome is equally distributed among both sexes despite racial variation.

Among the numerous manifestations of this syndrome, the vascular manifestation is most frequently observed as port wine birthmarks that affect the skin and face. Similar vascular malformations that involve the brain are referred to as leptomeningeal angiomatosis, which predominantly occurs on the same unilateral side as the birthmarks.[9] Common neurological symptoms include seizures, strokes, visual deficits, calcifications, and cerebral atrophy. Stroke episodes are commonly observed after the age of 5 and are treated with antiepileptic drugs, while migraines and headaches predominantly affect adults.[10]

Capillary malformations, commonly referred to as port wine stains, are one of the most frequently observed types of vascular malformations. They are often observed in the temple region, eyelid, and forehead, and they are noted to have a characteristic and distinct pattern. The gold standard light-based technology utilized for treatment thereof is a pulse diode laser. Notable ocular involvement with glaucoma is reported in 30.0%–70.0% of cases.[7] Topical and systemic pharmacotherapy are mostly used as primary management, whereas surgical and laser therapy are suggested when glaucoma is not brought under control.

Diagnosis becomes challenging when cases only report port wine birthmarks with no neurological symptoms, thereby relying on clinical and radiological imaging **(Figure 4).** One such instance is our case, which presented with only a port wine birthmark, a history of glaucoma, and an intraoral angioma with no other neurological symptoms and negative imaging findings. Magnetic resonance imaging plays an important role in ruling out any underlying neurological issue, such as effectively detecting leptomeningeal enhancement, abnormal venous drainage, and cortical atrophy. Susceptibility-weighted imaging and fluid-attenuated inversion recovery are powerful techniques that provide exceptional sensitivity in detecting pial vascular abnormalities. Their ability to reveal these subtle changes makes them invaluable tools in modern neuroimaging. Computed tomography aids in identifying structural abnormalities, such as the presence of any tram track calcifications and cortical atrophy. Moreover, positron emission tomography and single-photon emission computed tomography offer valuable insights for patients with Sturge–Weber syndrome by detecting subtle metabolic and perfusion changes.[7]

In recent times, targeted therapy has paved the way in managing Sturge–Weber syndrome with drugs such as Sirolimus[11] and topical rapamycin.[12] Gingival hyperplasia and angiomatosis that involve

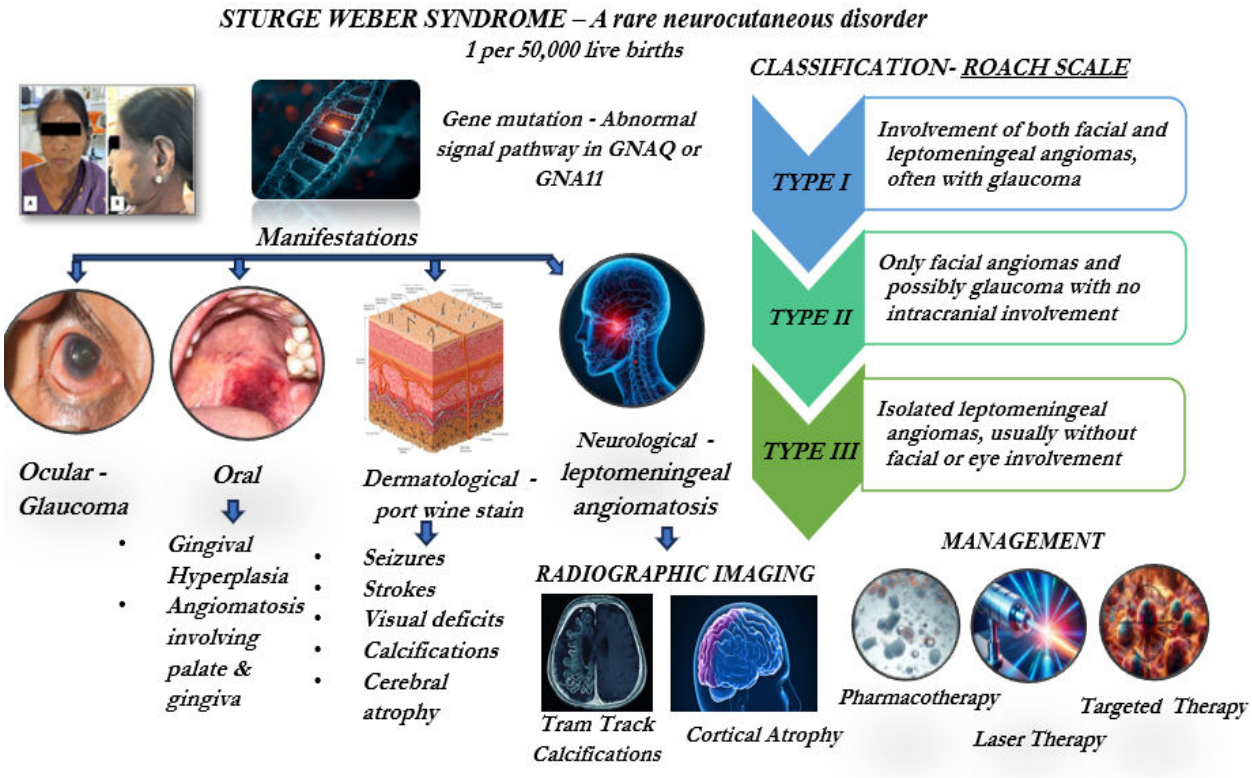

Figure 4. The clinical spectrum of Sturge–Weber syndrome.

the palate and gingiva are commonly reported intraoral findings.[13] Cases associated with mental retardation are often diagnosed with poor oral hygiene and gingival overgrowth as a complication of anticonvulsant drug use. The impact of even minimal trauma can lead to gingival bleeding in these cases. Patient education and preventive measures should be of paramount importance when planning any dental procedures.[3] The use of 0.1% chlorhexidine solution has been shown to be effective in controlling plaque biofilms affecting the periodontium in cases with Sturge–Weber syndrome.[2, 14] Over-instrumentation in endodontic cases is to be avoided to prevent hemorrhagic risk. Furthermore, CO_2 laser therapy has been proven successful in the management of gingival hyperplasia, thereby avoiding hemorrhage.[15] Hence, when planning a dental treatment strategy for Sturge–Weber syndrome cases, special precautions need to be taken to avoid post and prehemorrhage risks.

Conclusion

The clinical course of Sturge–Weber syndrome is highly unpredictable, with the prognosis primarily depending on the specific manifestations that are present. Cases classified as type II disorders are particularly challenging and require a multidisciplinary approach for prompt diagnosis. The port-wine birthmark, which is a prominent feature of this syndrome, can affect patients throughout their lives, thereby potentially diminishing their quality of life and leading to severe psychological trauma and a lack of self-confidence. Ongoing monitoring of individuals with these disorders is essential to assess disease progression over time. Clinicians need a solid understanding of the clinicopathological and radiological aspects to facilitate early diagnosis at the grassroots level.

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Conflict of interest

The authors declare that they have no conflicts of interest.

Data sharing statement

Data generated or analysed for the present report are included in this published article. Further details are available from the corresponding author on reasonable request after deidentification of the patient whose data are included in the report.

References

1. Robert J. Gorlin M. Cohen M, Raoul CM. Hennekam syndromes of the head and neck, New York: Mcgraw-Hill Book; 1964. Chapter: Hamartoneoplastic Syndromes P. 456–9.
2. Fortuna T, de Oliveira CA, Medrado AR, de Oliveira VM, Tunes RS, Marchionni AM. Dental care challenges in Sturge-Weber syndrome: a case report. Rev Ciênc Méd Biol 2020;19:642–6.
3. Nerkar A, Gadgil R, Bhoosreddy A, Shah K. Hemorrhagic risks associated with dental procedure in a rare case of type II Sturge-Weber syndrome. Inter J Cur Res Rev 2017;9:21–5.
4. Neto FX, Junior MA, Ximenes LS, de Souza Jacob CC, Junior AG, Palheta AC. Clinical features of Sturge-Weber syndrome. Intl Arch Otorhinolaryngol 2008;12:565–70.
5. Wyllie E, Gupta A, Lachhwani DK, editors. The treatment of epilepsy: principles and practice. Philadelphia: Lippincott Williams & Wilkins; 2006.
6. Mapara PN, Taur SM, Hadakar SG, Devendrappa SN, Gaonkar NN, Gugawad S, et al. Sturge–Weber syndrome: roots to a cure a nightmare in pediatric dentistry. Int J Clin Pediatr Dent 2021;14:145–8.
7. Naja K, Kapciak A, Górny J, Świerzyńska A, Hawajski A, Jankowska N, et al. Sturge-Weber Syndrome: a comprehensive review of clinical features, optimized diagnosis and management strategies. Qual Sport 2024;36:56849.
8. Nguyen V, Hochman M, Mihm Jr MC, Nelson JS, Tan W. The pathogenesis of port wine stain and Sturge Weber syndrome: complex interactions between genetic alterations and aberrant MAPK and PI3K activation. Int J Mol Sci 2019;20:2243.
9. Comi AM. Presentation, diagnosis, pathophysiology, and treatment of the neurological features of Sturge-Weber syndrome. Neurologist 2011;17:179–84.
10. Sánchez-Espino LF, Ivars M, Antoñanzas J, Baselga E. Sturge-Weber syndrome: a review of pathophysiology, genetics, clinical features, and current management approache. Appl Clin Genet 2023;16:63–81.
11. Freixo C, Ferreira V, Martins J, Almeida R, Caldeira D, Rosa M, et al. Efficacy and safety of sirolimus in the treatment of vascular anomalies: a systematic review. J Vasc Surg 2020;71:318–27.
12. Triana Junco PE, Sánchez Carpintero I, López Gutiérrez JC. Preventive treatment with oral sirolimus and aspirin in a newborn with severe Sturge Weber syndrome. Pediatr Dermatol 2019;36:524–7.
13. Tripathi AK, Kumar V, Dwivedi R, Saimbi CS. Sturge-Weber syndrome: oral and extra-oral manifestations. BMJ Case Rep 2015;2015:bcr2014207663.
14. Shaikh SM, Goswami M, Singh S, Singh D. Sturge–Weber syndrome–A case report. J Oral Biol Craniofac Res 2015;5:53–6.
15. Darbar UR, Hopper C, Speight PM, Newman HN. Combined treatment approach to gingival overgrowth due to drug therapy. J Clin Periodontol 1996;23:941–4.