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Case Report Open Access

Sturge Weber Syndrome Type I: Case Report and Literature Review

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Abstract

Sturge Weber Syndrome is caused by a somatic mosaic mutation in GNAQ gene leading to capillary malformations. It is characterized by port-wine stain, leptomeningeal angioma, glaucoma, seizures and mental retardation. We present a case of Sturge Weber Syndrome from Nepal. We also have emphasized on the review of literature of other reported cases of this syndrome from Nepal.

Keywords Sturge Weber Syndrome; Port wine stain; Leptomeningeal angiomas; Glaucoma; Rare syndrome Nepal

Introduction

Sturge Weber Syndrome also known as Encephalotrigeminal angiomatosis or Sturge Weber angiomatosis was first described by Schirmer in 1869. Later, William A. Sturge reported facial naevus in a 61/2 year old girl in 1879 and in 1897 Kalischer indicated cerebral involvement. It was in 1922 that Frederick Parkes Weber for the first time recorded the intracranial calcifications in a patient presenting with features similar to that described by Sturge and Kalischer [1-3].

In this article we are reporting a case of Sturge Weber Syndrome, which is most likely the third case to be reported in a male patient out of five cases that have been published from Nepal so far.

Case Report

We are describing here a case of Sturge Weber Syndrome seen in a 30 year old male patient, resident of Tarai area of Nepal. He came with the complaint of swelling of lips and gums since 4 years. He was mentally subnormal and unable to speak since birth. The gait was unusual. There was a history of occasional nasal bleeding and the lip swelling was also associated with bleeding on slight touch. Swelling was reported to have gradually increased in size over the period of 4 years. The patient's birth had been at full term by normal delivery but he had blurred vision, hearing disability, occasional epistaxis and history of seizures since birth. The family history did not reveal any similar complaints from his siblings. Extraoral examination revealed swelling and hemangioma of the lips. Swelling was asymmetrical on upper right side and was extending uniformly over the lower lip. Port wine nevus was present over the face bilaterally (Figure 1).

Palpable, movable submandibular lymph nodes were detected bilaterally. Intraoral examination disclosed severely compromised oral hygiene along with hemangiomatous proliferation of gingiva in both lower and upper anterior region. Hemangioma was seen extending bilaterally over the hard palate. On palpation gingiva was soft to firm in consistency. Severe malocclusion along with multiple carious teeth and root stumps was observed. Few teeth were missing (Figure 2).

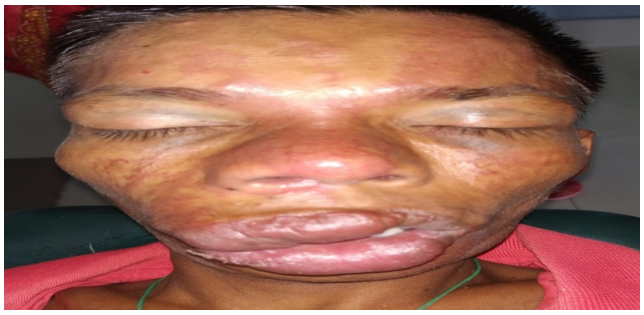

Figure 1: Post wine nevus present bilaterally over the face. Swelling involving both the lips and also present over right half of the nose.

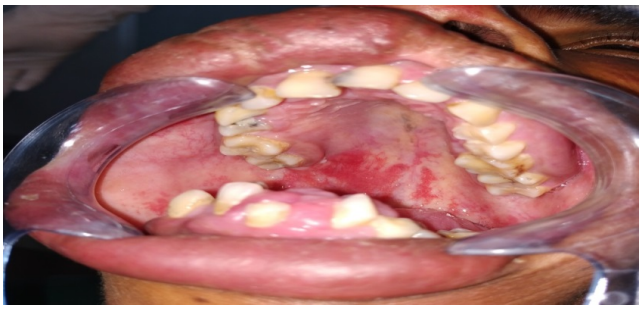

Figure 2: Hemangiomatous proliferation of gingiva along with malocclusion. Hemangioma present bilaterally, over the hard palate.

Ophthalmic examination revealed posterior subcapsular cataract in right eye. Digitally intraocular pressure was elevated in both the eyes. A diagnosis of bilateral glaucomatous optic atrophy was made and the patient was prescribed Timolol 0.5% -BE; 1 drop twice daily. CT scan of head showed atrophy of the left frontal and parietal lobes with prominence of adjacent sulci. No calcific areas or increased vascularity was noted in the region (Figure 3). Final impression of left

sided hemiatrophy of frontoparietal lobes was made.

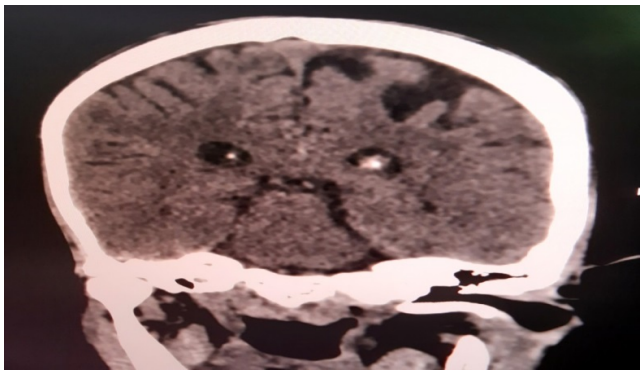

Figure 3: Atrophy of left frontal and parietal lobes. No calcific areas are evident.

Results and Discussion

Sturge Weber Syndrome is considered to be a rare, sporadic, non-hereditary, congenital, neurovascular disorder. They are a group of disorders collectively known as phakomatoses which involve neuroectodermal and at times mesodermal abnormalities as well [1]. The genetic cause accounting for it is a p.R183Q somatic mosaic mutation in GNAQ gene located on chromosome 9q21.2. The gene encodes Gαq, a guanine nucleotide protein. Mutation in this gene results in capillary venous malformations in the brain that presents itself with ophthalmic, neurological and cognitive symptoms that may or may not be associated with one another [4,5].Literature review shows that numerous theories have been put forward for the pathogenesis of this syndrome but the magnitude to which these changes affect the outcome in Sturge Weber Syndrome is unclear [6] and the most likely pathogenesis is linked with somatic activating mutations in GNAQ (alpha subunit q of the guanine nucleotide-binding protein heterodimer) gene. The mutation in GNAQ has been predicted to cause activation of downstream pathway contributing to port wine stains and syndromic characteristics seen in Sturge Weber Syndrome patients [6-8].The clinical features in Sturge Weber Syndrome vary extensively and may include cutaneous, neurologic, ophthalmologic and oral manifestations. Differentials that need to be considered for this syndrome includes Rendu Osler Weber syndrome, Parkes Weber syndrome, CLOVES syndrome and Klippel-Trenaunay-Weber syndrome which were ruled out based on the clinical and radiographic features in the present case [9].

Based on Roach's scale that classifies the encephalofacial angiomatosis, Sturge Weber Syndrome has clinically been divided into 3 types (Table 1) [10].

Type	Facial angioma	Leptomeningeal angioma	Glaucoma
1[a]	+	+	+/-
2	+	-	+/-
3	-	+	-/+b

[a] Classic Sturge-Weber-Syndrome

[b] Usually not present

Table 1: Roach's scale for classification for types of Sturge-Weber Syndrome

On basis of Roach's scale the present case was put under Type I category, Classical Sturge Weber Syndrome. To the best of our knowledge five cases of Sturge Weber Syndrome have so far been reported and published from Nepal. Out of the 5 cases reported, 2 were males and all 5 cases presented with Facial angioma and almost all had glaucoma. The age, sex and the clinical features are listed in Table 2 [11-15] and neuroradiological findings described in these 5 previously reported cases.

Year	Authors	Age/Sex	Features
2004 [11]	Kumar V, Prasad BK	8 year/Male	Mental Retardation, Deep purple nevus, X-ray skull showed calcification with focal atrophy
2010 [12]	Devkota S, Upadhyay S	New born/Female	Single episode of Partial seizure, Dark pink to red port wine stain in right half of face. Bilateral congenital glaucoma
2013 [13]	Singh P, Singh S	17 year/Female	Mental Retardation, Convulsons, Glaucomatous optic atrophy, Hypervascularity of right side of palate, soft palate and buccal mucosa, hemangioma of upper lip, Intracranial calcification I n occipital lobe extending to parietal lobe
2013 [14]	Thapa R, Shields CL	17 year/Male	Blurred vision, Left eye showed periocular nevus flammeus, Diffuse choroidal hemangioma
2015 [15]	Lavaju P, Mahat P	6 year/Female	Bilateral Glaucoma, Port wine stain involving both side of face, right ear lobule and right side of upper trunk with mild right sided facial hypertrophy

Table 2: List of details of 5 previously reported cases from Nepal.

The treatment and prognosis in such patients depend entirely on the type and severity of the features. Management becomes challenging because of increased chances of hemorrhage and attaining hemostasis can be troublesome. Most effective treatment procedure in such conditions is considered to be gingivectomy along with use of LASERs that can help to achieve hemostasis [9]. In present case we

lost the follow-up with the patient, as he was unable to come later due to COVID-19 pandemic.

Conclusion

We emphasize that more awareness and profound knowledge of this rare syndrome should be there among the oral health care practitioners so that on one hand there can be an early screening and inter departmental consultations, while on the other hand any untoward life threatening complications can be avoided.

Declaration of patients Consent

Written informed consent was obtained from the patient's guardian for publishing this case report and accompanying images. A copy of the written consent is available for review by the editor-in-chief of this journal.

Disclosure

The authors report no conflict of interest in this work.

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