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Case Report

Oncology Section

An Unusual Case of Sturge-Weber Syndrome with Squamous Cell Carcinoma of Tongue

RAJDEEP GUHA[1], SUKANYA NASKAR[2], SUBHABRATA GHOSH[3], ANANNYA TRIPATHY[4], ARNAB ADAK[5]

ABSTRACT

Sturge-Weber Syndrome (SWS) is a rare congenital neurocutaneous syndrome affecting skin and soft tissues in the distribution of cranial nerve V, the ipsilateral cerebral hemisphere, and eye, with an incidence of 1 in 20,000 to 50,000 births, with no overall predisposition to developing cancers. There is a dearth of literature for the use of microvascular free flaps in SWS, and none pertaining to its use in oncological surgery for these patients, to the best of our knowledge. Free flaps have been used to reconstruct port wine stains in such patients. Hereby, authors present a case of a 52-year-old male having SWS who developed a Squamous Cell Carcinoma (SCC) of the tongue and was managed surgically, with the reconstruction being done with a free Radial Artery Forearm Flap (RAFF). The patient complained of ulcer over tongue since three months. He was already diagnosed case of SWS. Patient's extraoral examination also revealed large hyperpigmented pinkish-brown patch with multiple nodules over the left half of his forehead, extending from hairline till the upper eyelid. The histopathology report showed a moderately differentiated SCC tumour with a maximum dimension of 3.2 cm, for which patient also received adjuvant radiotherapy. This case report demonstrates the safe use of free flaps in patients with head and neck cancers who have a history of SWS.

Keywords: Carcinoma, Free flap, Neurocutaneous syndrome, Tongue neoplasms

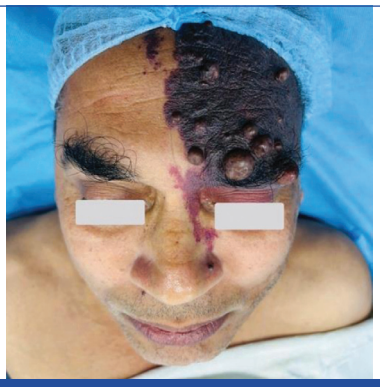
CASE REPORT

A 52-year-old male presented to the head and neck surgical Outpatient Department (OPD) with complaints of an ulcer over the right side of his tongue, since the last three months. The ulcer was gradually progressive and painless, not associated with bleeding, difficulty in swallowing or speech, or any neck swellings. He had a habit of beedi smoking for ten years, which he stopped since the last two years ago. He denied the consumption of alcohol or smokeless tobacco and had no associated comorbidities.

The patient was a diagnosed SWS since childhood and was on regular antiepileptic medications for seizure control, with the last seizure occurring six years prior to presentation. He was taking oral Divalproex extended-release tablets (500 mg) twice daily for the last five years, with good seizure control.

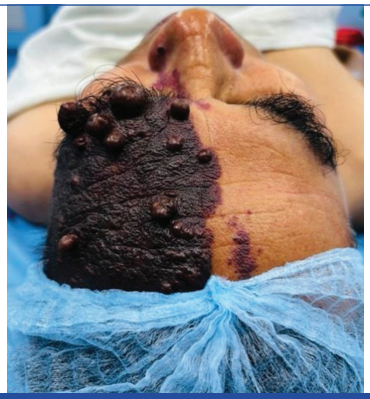
On examination, the patient was well-oriented and well-nourished, with normal intelligence and an Eastern Cooperative Oncology Group (ECOG) performance status score of 1.

There was presence of a large hyperpigmented pinkish-brown patch with multiple nodules over the left half of his forehead, extending from hairline till the upper eyelid; however, the eye itself was uninvolved, with visual acuity of 6/6. As per the Roach Classification, he had Type I (classical) SWS [Table/Fig-1,2] [1].

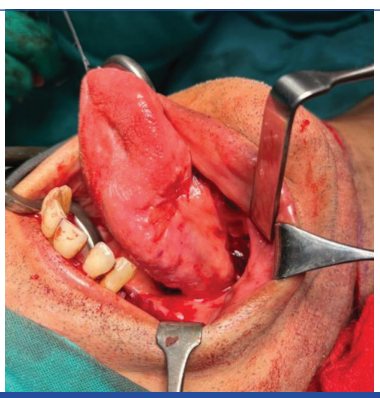


[Table/Fig-2]: Frontal view of the lesion.

On intraoral examination, the mouth opening was adequate, with an ulcero-proliferative growth of approximate size 3×1 cm present over the right lateral border of the tongue, extending 2 cm away from tip, with a posterior extent upto 2nd molar. The midline, base of tongue, and floor of mouth were uninvolved, with no restriction of tongue movements. There was no palpable cervical lymphadenopathy, classified as Tumour (T)2, Lymph Node (N)0, Metastasis (M)0 as per American Joint Committee on Cancer (AJCC) 8th edition [Table/Fig-3] [2].

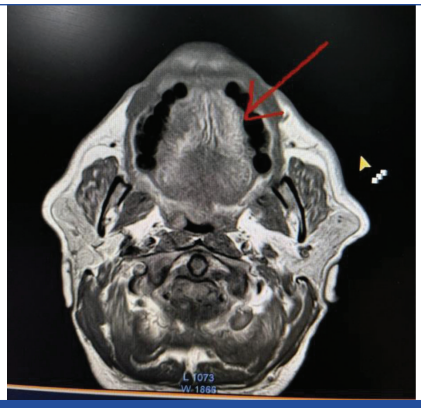


[Table/Fig-1]: Facial port wine stain and angiomatosis superior view.



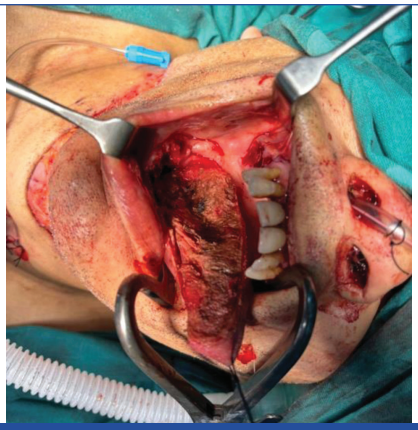
[Table/Fig-3]: Intraoral view of the tongue lesion.

The patient underwent all routine investigations, along with a contrast-enhanced Magnetic Resonance Imaging (MRI) of the face and neck, plus a Contrast Enhanced Computed Tomography (CECT) scan of the thorax as part of his work-up. A punch biopsy was taken from the lesion, which was suggestive of moderately differentiated SCC. The MRI showed hyperintense lesion measuring 31×19×16 mm in left anterior 2/3rd of tongue, with patchy contrast enhancement, involving the genioglossus muscle. The mandible was uninvolved, with sub-centimetric nodes at level Ia and right level III. CT thorax showed no abnormal lesions [Table/Fig-4].



[Table/Fig-4]: Contrast enhanced MRI scan demonstrating the tumour. (T1 weighted image, with contrast).

The patient was managed surgically with a right hemiglossectomy, right-sided extended supra-omohyoid neck dissection, left-sided supra-omohyoid neck dissection, tracheostomy, and reconstruction with a left free RAFF under general anaesthesia. The neck dissection was done via a transverse skin crease incision, and the primary lesion was excised via an intraoral approach [Table/Fig-5].



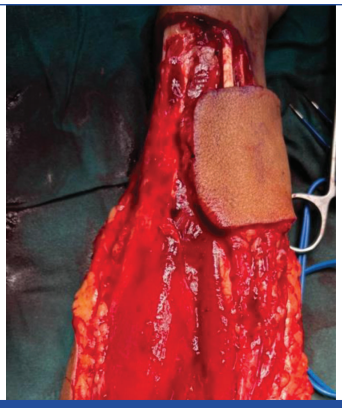
[Table/Fig-5]: Defect following surgical excision of tumour.

Simultaneous harvesting of the RAFF was done and insetting of flap was done with microvascular anastomosis of left radial artery with right facial artery, while left cephalic vein and vanae comitantes were anastomosed end-to-side with the right internal jugular vein. The donor site was closed with a Split Thickness Skin Graft (STSG) harvested from the left thigh [Table/Fig-6].

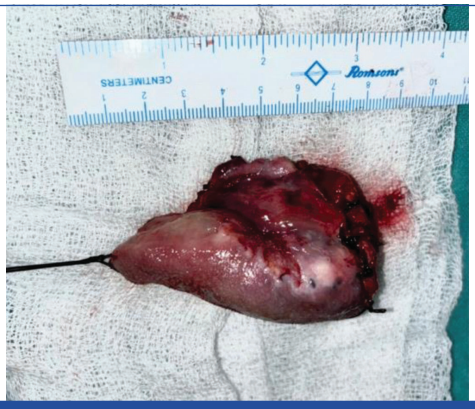
The resected specimen obtained from hemiglossectomy revealed a well-demarcated lesion involving the lateral tongue [Table/Fig-7].

The patient had an uneventful postoperative period and was decannulated and discharged on Postoperative Day (POD) 5, with a healthy flap [Table/Fig-8]. Donor site dressing was changed, and good uptake of the STSG was noted. He underwent Ryle's tube feeding and routine speech and swallowing rehabilitation and was allowed oral sips of water on POD 7, with soft diet started on POD 10.

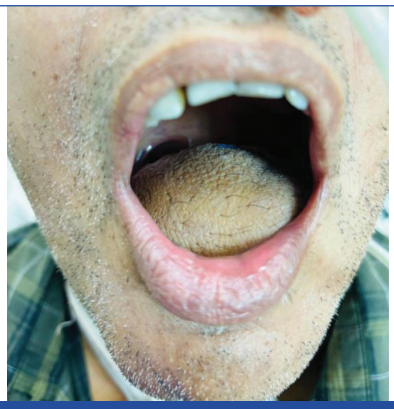
Patient was followed-up on POD 20 and was on full oral diet, with good healing of neck, thigh, and hand wounds. The final histopathology report showed a Moderately Differentiated Squamous Cell Carcinoma (MDSCC) tumour of maximum dimension 3.2 cm,



[Table/Fig-6]: Radial artery forearm flap harvested, prior to division of vascular pedicle.

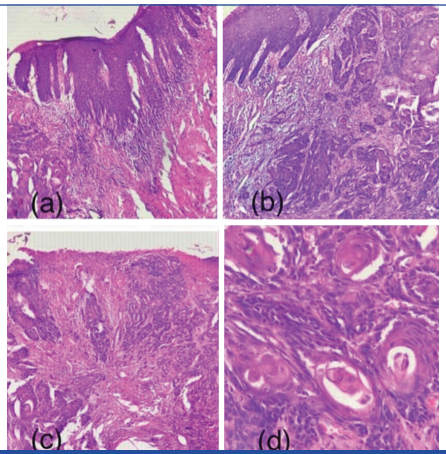


[Table/Fig-7]: Resected surgical specimen of hemiglossectomy.



[Table/Fig-8]: Satisfactory flap uptake as seen on Postoperative Day (POD) 5.

with all resection margins free; a Depth of Invasion (DOI) of 9 mm; and Worst Pattern of Invasion (WPOI) of 4. Lymphovascular invasion was absent; however, perineural invasion was present [Table/Fig-9]. 3/74 ipsilateral nodes were positive for malignancy, without



[Table/Fig-9]: Histopathological images of moderately differentiated squamous cell carcinoma of the tongue stained with H&E. a-c) showing invasive epithelial islands (10X magnification); d) showing keratin pearls and nuclear atypia (40X magnification).

extra-nodal extension (pT2N2b). Based on the above, the patient underwent adjuvant radiotherapy, completed the same, and has a Disease-Free Interval (DFI) of seven months on follow-up.

DISCUSSION

Sturge-Weber Syndrome (SWS) is a congenital neurocutaneous syndrome affecting the brain, eyes, and skin in the distribution of the V1 division of the trigeminal nerve, occasionally involving the V2 and V3 divisions as well [3,4]. It is a sporadic disorder characterised by facial and intracranial capillary and vascular malformations. Patients typically present with a facial nevus (port-wine stain), usually unilateral and non progressive, alongside infantile seizures. Other features include eye enlargement, glaucoma, choroidal haemangioma, maxillary hyperplasia, and intellectual disability [3,5].

The SWS affects roughly 1 in 20,000 to 50,000 births, occurring as a consequence of a somatic mutation in the guanine nucleotide-binding protein G (q) (GNAQ) gene [4,5]. However, SWS is not associated with an increased risk of cancer development compared to other neurocutaneous syndromes like Von Hippel-Lindau Syndrome. The main clinical problems of SWS are due to the intracranial vascular abnormalities [6,7].

Hereby, authors, have reported the case of an adult with SWS who developed a SCC of the oral tongue and was treated surgically, with the ablative defect reconstructed using a free flap. There is a dearth of literature for the use of free flaps in SWS, and none pertaining to its use in cancer surgery for these patients, to the best of our knowledge. Free flaps have been used with various degrees of success in the management of facial port-wine stains in SWS [8,9].

The SWS (encephalotrigeminal angiomatosis) is itself a rare entity, however, in countries like India, which have high rates of tobacco smoking and smokeless tobacco consumption, encountering an adult SWS patient with head and neck SCC may pose a challenge that any head and neck surgeon could potentially come across.

There are reports in literature of microvascular anastomosis being used successfully in other neurocutaneous syndromes. For instance, a three-year-old child with tuberous sclerosis was successfully treated for a fast growing brachial aneurysm with a microsurgical reconstruction using reversed greater saphenous vein graft [10]. In a series of six patients by Hu L et al., it was demonstrated that anterolateral thigh and latisimus dorsi free flaps can be used to cover surgical defects due to disfiguring giant facial neurofibromas in patients with Neurofibromatosis Type I (NF-1) [11].

A similar series from Uygur F et al., showed the use of microvascular free flaps in five patients with NF-1, in which all the free flaps survived; however, patients had recurrent tumours [12]. In the case of our patient, there was no difference in the wound healing or perioperative complications during free flap procedure, suggesting that it may be considered a viable option for reconstruction in well-selected patients. A thorough history, pre-anaesthetic work-up, and clearance from a neurologist should first be obtained due to the risk of seizures and the prolonged nature of surgery, in order to reduce perioperative morbidity.

CONCLUSION(S)

Microvascular free flaps are a viable and safe option for reconstruction of surgical defects due to oncological surgery, SWS lesions existing in the head and neck region and in well-selected patients, the outcomes are favourable.

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PARTICULARS OF CONTRIBUTORS:

1. Specialist Grade I, Department of Head and Neck Surgical Oncology, Chittaranjan National Cancer Institute, New Town, Kolkata, West Bengal, India.
2. Specialist Grade II, Department of Head and Neck Surgical Oncology, Chittaranjan National Cancer Institute, New Town, Kolkata, West Bengal, India.
3. Consultant, Department of Head and Neck Surgical Oncology, Chittaranjan National Cancer Institute, New Town, Kolkata, West Bengal, India.
4. Fellow of National Board, Department of Head and Neck Surgical Oncology, Chittaranjan National Cancer Institute, New Town, Kolkata, West Bengal, India.
5. Resident, Department of Surgical Oncology, Chittaranjan National Cancer Institute, New Town, Kolkata, West Bengal, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Anannya Tripathy,
1st Floor, 35/9C, Labanya Apartments, 214, P Mazumdar Road, Purbanchal Canal South Road, Kolkata-700078, West Bengal, India.
E-mail: anannyatripathy14@gmail.com

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