

Sturge-Weber syndrome with massive macroglossia and anterior neck space infection- a case report and review of literature

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Abstract

Sturge-Weber syndrome (SWS), is a rare neuro-cutaneous angiomas which affects male and females alike. The clinical manifestations include angiomas, haemangiomas of the lips, tongue and palatine region. The oral manifestations are usually unilateral and are susceptible to bleed. Patients can also present with macroglossia and maxillary bone hypertrophy which can lead to malocclusion of the oral cavity. Food accumulation due to occlusion can cause growth of bacteria which can intensify infections and can cause gingival hyperplasia. A case of a middle-aged 39 year old female was reported in the Ziauddin Hospital, Karachi on 2nd of February, 2022 with the presenting complaints of intermittent fever and drowsiness for 10 days. On examination she had massive tongue enlargement, drooling, malocclusion, difficulty in eating and breathing. She was a known case of Sturge-Weber syndrome. Based on the clinical and radiological findings, she was managed along the lines of pre-laryngeal soft tissue and submandibular infection.

Keywords: Anterior neck space infections, Macroglossia, Sturge weber syndrome, Neurocutaneous disorder, Rare presentation in sturge weber.

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Introduction

Sturge-Weber syndrome (SWS) is a rare neurocutaneous disorder which belongs to the group phakomatoses. It is also known as encephalo facial angiomas or cephalotrigeminal angiomas. It affects males and females alike and has no racial predisposition¹. The angiomas affect, the leptomeninges and the skin of the face and choroid, typically in the distribution of V1 and V2 of the Trigeminal nerve. Other clinical manifestations include haemangiomas of the lips, tongue and palatine region². The oral manifestations are usually unilateral and

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are susceptible to bleed. Patients can also present with macroglossia and maxillary bone hypertrophy which can lead to malocclusion of the oral cavity. The gingival hyperplasia is often the most common oral manifestation. Food accumulation due to occlusion can cause growth of bacteria which can intensify infections and can cause gingival hyperplasia³. A case of SWS is presented who had progressively worsening macroglossia over years and was admitted with anterior neck space infection.

Case Report

A 39 year old female reported to the tertiary care teaching hospital (Ziauddin hospital, Karachi) on 2nd of February, 2022 with the presenting complaints of intermittent fever and drowsiness for 10 days. Her past medical history was of SWS on the basis of port-wine stain, glaucoma, intractable seizures and right tram-track calcifications on CT-Brain scan. There was a history of delayed developmental milestone and learning disabilities. She also had massive tongue enlargement and complaints of drooling, malocclusion, difficulty in eating and breathing. The swelling had been present for a very long time and had increased over last 15 years but there was no reported complaint of sudden recent increment in size of swelling, pain or acute compromised airway. Her breathing was not compromised while sitting or lying down but there was element of OSA.

On examination, patient was drowsy but arousable. Chest auscultation revealed bilateral basilar coarse crepitations, abdomen was soft and non-tender but distended. Extra oral examination showed the presence of port wine stain bilaterally along the ophthalmic division of the trigeminal nerve and ipsilateral congenital glaucoma. Intraoral examination could not be done because of the hypertrophy of the tongue. Major airways appeared patent (Figure 1 and 2).

On arrival, vitals were BP- 104/70, HR- 87/min, Saturation- 87% on Room Air. 1L of oxygen support was given via nasal prongs which were later tapered off within 24hrs. ABGs were done and there was no retention of CO₂.

CT scan of head and neck was done which showed evidence of significant inflammatory fat stranding within subcutaneous tissue at bilateral submandibular location



Figure-1: Lateral view of hypertrophied tongue.



Figure-2: Anterior view of hypertrophied tongue.

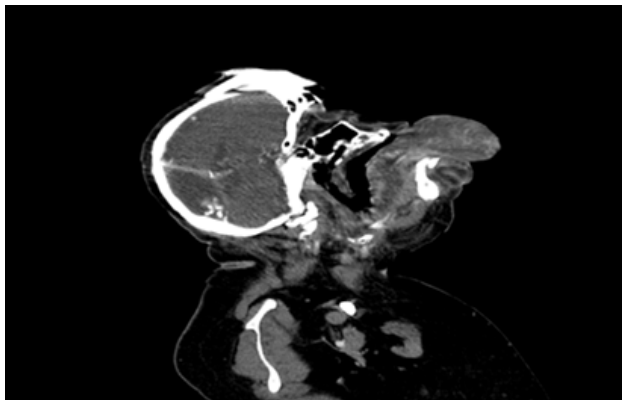


Figure-3: Sagittal image showing gross glossomegaly with protruded tongue.

and pre-laryngeal soft tissue. Few enlarged and enhancing lymph nodes were also identified at neck predominantly at bilateral level I & II. Prominent vascularity was also appreciated within the left upper cheek region. Muscular hypertrophy and enlargement of tongue was noted with significant protrusion. There was no definite narrowing of upper airway. Sections through the brain showed hemiatrophy of right cerebral hemisphere along with dense areas of cortical and subcortical location in right cerebral hemisphere

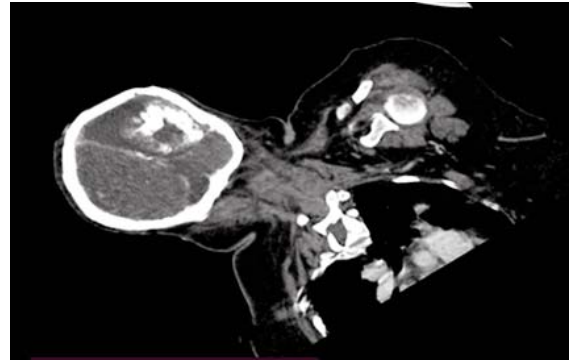


Figure-4: Coronal image showing sub cortical and cortical marked calcification with loss of brain parenchyma suggesting Sturge Weber.

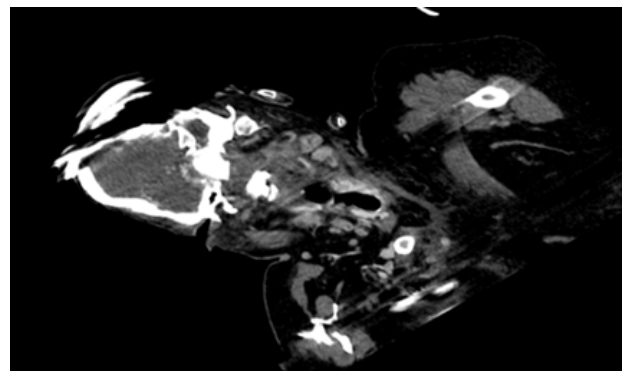


Figure-5: Coronal image showing fat stranding and lymphadenopathy in anterior triangle of neck.

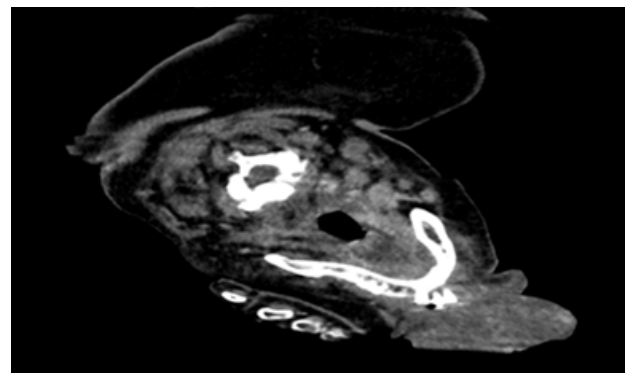


Figure-6: Axial image showing fat stranding and lymphadenopathy in anterior triangle of neck.

suggestive of SWS. Focal calcification was seen along the posterior chamber of right eye globe representing haemangioma as seen in case of SWS. Right ethmoid and right maxillary sinusitis were also seen. (Fig 3-6)

CT scan of the chest did not reveal any signs of pulmonary infection. On admission baseline investigations with additional tests were done to ascertain the source of infection. Admission labs showed increased infective

markers (CRP474mg/dl – n: <5mg/dl), TLC count of $13 \times 10^9/L$ (n= 4- $10 \times 10^9/L$) and Blood cultures showed no growth. She was started on Piperacillin-Tazobactam 4.5gm thrice daily to which she responded. Her CRP dropped to 240mg/dl and her TLC count decreased to $10 \times 10^9/L$. Her LFTs were deranged likely due to antiepileptic (Valproate) long term use. Neurologist was taken on board and thought that the drowsiness was due to Encephalopathy likely toxic(infective) /metabolic entity (Valproate). In the context of Valproate use, CLD was suspected and Ammonia levels were sent which were elevated 95umol/L (normal range 10-47). Hence, Valproate was stopped and Levetiracetam and Carbamazepine were continued. Lactulose was advised to decrease ammonia levels. Patient's clinical condition was improved within 48hrs. As, there were no risk factors for MRSA or Pseudomonas on the initial presentation therefore, antibiotics were later switched to oral Co-amoxiclav 1gm bd and patient was discharged on family's request.

She was readmitted after 3 days with drowsiness and this time the admission period was 10 days. On arrival to the ER – BP 130/90 - Pulse 98 - Saturations 96% on room air and temperature was 102°F. On examination, the patient was drowsy and her chest had conducting sounds bilaterally. There was mild abdominal tenderness in the epigastric region and throat secretions which needed regular suctioning. All baseline investigations were sent which showed elevated WBC and CRP from before. The possibility of untreated previous infection vs Cather related UTI and HAP was entertained. New chest x-ray showed no definite consolidation. Urine DR was normal. Previous admission urine and blood cultures showed no growth. Maximum doses Meropenem (gram negative coverage) and Teicoplanin (gram positive coverage) were started. Her fever persisted after 72hrs and CRP kept rising so Colistin was added. Sputum CS showed growth of Acinetobacter (only sensitive to Colistin). Echocardiography showed no evidence of endocarditis (presuming pre-laryngeal soft tissue infection). EF was 60% and no valvular abnormality was reported. Maxillofacial team was taken on board for the possible need of tracheostomy if the patient's condition deteriorates, and general assessment regarding the neck space infection. They advised to insert an NG tube for feeding but the family did not give consent. The team agreed with antibiotics and did not advise any further intervention. Insertion of CVP line was requested but consent was not given for this. Gradually her fever settled. Her consciousness and responsiveness also got better. Hence, the patient got discharged with an advice to complete the course of antibiotics for 14 days and follow-

up with repeat inflammatory markers.

Based on the history, clinical and radiological investigations, a diagnosis of pre-laryngeal soft tissue and sub mandibular infection (secondary to possible dental infection due to obstruction by the hypertrophied tongue). Divalproex induced hepatic dysfunction causing elevated ammonia were thought to be due to deranged liver function tests as no other reason could be specified.

A thorough plaque control regimen was suggested to reduce occurrence of dental infections. Options of glossectomy and fenestrated tracheostomy were suggested but family were not in favour of any surgical intervention. They were an educated family and were very devoted to the wellbeing of their relative and were aware of the risks and benefits. Weekly follow-up of the infective markers showed a declining pattern after which her antibiotics were stopped.

Discussion

Sturge-Weber syndrome is the third most common neurocutaneous disorder after tuberous sclerosis and neurofibromatosis. It is characterized by angiomas in leptomeninges, face and choroid. The mutation that leads to this is caused by GNAQ gene in the long arm of chromosome⁹. The facial malformation is manifested as port wine stain.⁴

Neurological manifestations include atonic, tonic and myoclonic seizures which are usually the first neurological manifestations. Furthermore, they can also be manifest as transient stroke, behavioural problems, hemianopsia and hemiplegia.⁵ Poor cognitive outcomes have been associated with early onset and high frequency of seizures and bilateral involvement of the brain. This suggests that preventing and controlling seizure is absolutely necessary in such patients.⁶

If the oral cavity gets involved then it can manifest as macroglossia, maxillary bone hypertrophy, oral mucosal haemangioma and tooth maleruption.⁵

Manifest ocular symptoms which are usually ipsilateral to the port wine stain, are seen in 50% of the patients. It can involve the eyelid, cornea, anterior chamber and retina. Glaucoma develops secondary to the anatomical alterations and the most common is the open angle glaucoma. It shows bimodal peak in which 60% of patients develop glaucoma in early childhood and 40% develop in late adolescence. A routine slit-lamp examination is usually sufficient to detect all kind of abnormalities.⁷

Kennedy Krieger Institute (KKI) implemented universal

suicide risk screening in all outpatient medical clinics in August 2017 who presented with SWS. The study showed that patients with SWS are more susceptible to suicidal ideations than patients with some other neurological abnormalities. Thus, families who have patients suffering from SWS should be counselled in detail regarding the risks of suicidal ideations⁸. The reported case of 38 year old female patient manifested all the symptoms attributed to SWS. However, she had massive macroglossia which is not commonly seen. She developed recurrent pre-laryngeal and submandibular infections secondary to occlusion. Her dental hygiene was very poor. So one should be aware of the importance of counselling patients to maintain a good oral hygiene amongst rest of the symptomatic treatment provided.

Conclusion

To conclude, the need for an appropriate and timely intervention in patients with Sturge weber syndrome is crucial due to the association of gingival vascular features and other complications such as infections.

Recommendation: Family of the patients diagnosed with SWS (Sturge weber syndrome) must receive appropriate counselling regarding maintenance of good oral hygiene and appropriate behavioural management in order to improve the quality of life.

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