Sturge-Weber syndrome: a case report with clinical and radiological features

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Abstract

Sturge Weber syndrome (SWS) is an uncommon, sporadically occurring, frequently progressive neuro-cutaneous syndrome, consisting of congenital hamartomatous malformations, often associated with epilepsy and variable oral manifestations. The port-wine nevus (PWN) is usually the first component of the syndrome, ocular and neurological manifestations like convulsions are among the most characteristic features of disease. Intra orally, the angiomatous lesions may involve the gingiva and buccal mucosa. Radiological investigations are most useful, with conventional radiography, computed tomography and magnetic resonance imaging playing a pivotal role in demonstrating the cerebral changes. This is a report of a case with oral involvement and highlights the importance of imaging in diagnosis and a multidisciplinary approach in treatment of SWS.

Key words: Sturge Weber syndrome, port-wine nevus, oral involvement, imaging modalities, tramline calcifications

Introduction

Sturge-Weber syndrome (MIM ID 185300) (ORPHA 3205) (ICD 10 code - Q85.8) is a developmental capillary vascular disorder originating during embryogenesis before neural crest migration, from errors in development of ectoderm and mesoderm in the anterior neural primordium due possibly to somatic mutations. It is characterised by facial capillary malformations (classically referred to as angiomas even though they are not tumours), accompanied by variable degrees of ocular and neurological anomalies. (Enjolras, 2006). The exact aetiology is unknown but the primary defect may be a developmental insult affecting precursors of tissues that originate in the promesencephalic and mesencephalic neural crest, which later give rise to vascular and other tissue malformations in the meninges, eye and the dermis. The orofacial manifestations are variable and include cutaneous facial PWN and ipsilateral hypervascular changes affecting the oral mucosa and gingiva. PWN is a dermal capillary vascular malformation present at birth (Thomas-Sohl et al., 2004). It is usually the first component of the syndrome to be noticed by patients. Invasive dental procedures involving affected sites in these patients may carry an increased risk of bleeding and hence dental practitioners should be prepared in case any complications ensue. Radiological investigations are most useful, with conventional skull radiographs, computed tomography and magnetic resonance imaging playing a pivotal role in demonstrating the cerebral changes. Treatment mainly consists of seizure control with anti convulsants. Surgery is reserved for refractory seizures, intractable glaucoma and laser therapy for facial nevus. Diagnosis and treatment of SWS relies on a detailed history, thorough clinical examination and imaging studies, signifying requirement of a multidisciplinary approach.

Case Report

A 24-year-old male patient reported to the dental department with a complaint of food lodgement and irritation of the tongue for the previous two months. A detailed dental history revealed that the patient had no complaint of premature tooth eruption or abnormal craniofacial growth. Medical history revealed that the patient had had epileptic seizures since the age of 10 and was under anticonvulsant therapy since then, with no recurrence over the past four years.

Extra oral examination revealed gross asymmetry of face with the nose deviated towards the right side and pinkish-red staining of left facial skin along the dermatomal involvement of all three divisions of trigeminal nerve. The pinkish-red stain extended superiorly from hairline to the line joining tragus of ear to angle of mouth inferiorly and medially from midline to 2cm in front of tragus of ear laterally. Increased choroidal vascularisation of left eye and left upper lip was evident (Figure 1). Intra oral examination revealed increased vascularity of left buccal mucosa (Figure 2), macroglossia, vertical crenations, horizontal fissuring and coated tongue (Figure 3). Gingival enlargement secondary to anticonvulsant drugs treatment is one of the commonly documented oral manifestations in patients SWS. Enlargement mainly affects the labial surface of the interdental papilla, though greater extensions can be affected including the gingival margins and lingual and palatal...
surfaces. The patient in focus did not present such gingival symptoms other than primary and secondary gingivomucosal involvement of intra oral angiomatous lesion. Maxillary midline diastema, maxillary anterior teeth spacing and proclination and increased overjet between maxillary and mandibular anterior teeth were some of the inter-arch discrepancies observed in the patient which could possibly have contributed to the macroglossia.

Panoramic radiography revealed an enlarged mandibular foramen and bifid inferior alveolar canal on the right side and a widened inferior alveolar canal on the left side which may signify abnormal draining veins (Figure 4). A brain CT scan revealed tramline calcifications in the frontal and parietal lobes of left side of the brain, partial hemiatrophy of the left cerebral hemisphere (Figure 5) and thickening of the skull vault involving frontal, parietal and temporal bones of the left side. Ocular examination revealed normal intra-ocular pressure of 18 mm Hg. Diascopy performed on the buccal mucosa produced blanching of mucosa on pressure application, suggesting a vascular nature of the lesion. A final diagnosis of Type 1 - Fronto Temporo Parietal SWS involving left side was given.

The patient was advised to perform regular tongue-scraping, a thorough plaque control regimen and use chlorhexidine mouth wash to avoid further risk of glossitis, inflammatory gingival enlargement and dental caries. Periodic follow up for every six months was advised. Orthodontic treatment was planned for the patient to correct the various intra and inter arch discrepancies. The patient was not willing to undergo any treatment for the facial port-wine nevus. A neurological consultation was undertaken, but no treatment was advised as the epileptic seizures were under control and no other neurological abnormality was detected.

Discussion

Figure 1. Pinkish-red staining of skin over the three divisions of left trigeminal nerve and labial angioma of left upper lip

Figure 2. Increased vascularity of left buccal mucosa

Figure 3. Macroglossia, vertical crenations and horizontal fissuring of dorsal surface of tongue

Figure 4. Enlarged mandibular foramen, bifid inferior alveolar canal on right side and widened inferior alveolar canal on left side
SWS also known as encephalo trigeminal haemangiomatosis, is an uncommon congenital condition. It is a sporadically occurring, frequently progressive, congenital neurocutaneous syndrome often associated with epilepsy which belongs to a group of disorders collectively known as ‘Phakomatoses’ or ‘Mother-spot disease’ but in contrast to the other disorders (Neurofibromatosis, Tuberous sclerosis and von Hippel-Lindau disease) in this group, there is no evidence of heredity. The possibility of a somatic mutation being present in SWS is based on the finding of an increased gene expression of fibronectin from port-wine stain (PWS) fibroblasts when compared to fibroblasts from normal skin in the same patient. The somatic mutation in the precursors of some angiogenic factors may lead to their over production, producing angiomas or due to a lethal gene surviving by mosaicism (Rajendran, 2006). SWS occurs with a frequency of approximately 1:50,000 live births. The influence of heredity is not documented.

The typical patient presents at birth with facial angiomas, however the reverse is not always true. In the incomplete form of Sturge-Weber syndrome, central nervous system angiomas occur without cutaneous manifestations, thus no suspicion of the syndrome arises until the onset of seizures (Arif et al., 2008). Angiomas of SWS result due to failure of regression of a vascular plexus around cephalic portion of neural tube which is destined to become facial skin. This vascular plexus normally forms at the 6th week of intrauterine life and regresses by 9th week. Failure of its regression results in residual vascular tissue which forms angiomatous malformations of leptomeninges, face and ipsilateral eye (Takeoka and Riviello, 2008). There may be seizures, developmental delay and mental retardation if angiomas involve a greater part of brain. Seizures of the contralateral side to the PWS may begin in infancy and worsen with age, eventually occurring in 75% cases. Mental retardation may be progressive but is not related to the seizure severity or frequency.

Patients affected with the syndrome present a variety of orofacial clinical manifestations including gingival haemangiomatous lesions, labial angiomas and nasal septum deviation (Tarsila et al., 2004). Macroglossia was present in our reported case, although it is not known to be associated with the syndrome. SWS is evident at birth as a unilateral PWS of the forehead and upper eyelid in the region supplied by the first branch of the trigeminal nerve, and varies in colour from pink to red to purple in colour: 50% of affected children will have or develop glaucoma. Glaucoma may occur when PWS involves the eyelids (Arif et al., 2008).

The diagnosis is based on clinical and imaging studies. Port wine nevus is observed clinically. Skull films may reveal tram track calcification caused by calcification in apposing gyri, ipsilateral calvarial thickening and enlargement of the para-nasal sinuses and mastoid. Cranial CT scan revealing cortical atrophy underlying the angioma with gyriform ‘tram-track’ calcifications is the characteristic imaging feature. MRI is the current gold standard for diagnosis of the disease which is reliable even in very young infants (Comi, 2007). Newborn babies with a PWS should have an ophthalmological examination in the first month of life, followed by neuroimaging (CT and gadolinium enhanced MRI) by 6-12 months age or sooner if neurologic signs are present. Cerebral blood flow imaging, Single Photon Emission Computed Tomography (SPECT) and Positron Emission Tomography (PET) are also useful when possible. The neurological signs are due to ipsilateral leptomeningeal angioma involving the occipital and posterior parietal lobes of the brain; vascular stasis with resultant ischaemia leads to calcification and laminar cortical necrosis.

Management of the syndrome involves both medical and surgical approaches. Medical treatment includes anticonvulsant therapy with prophylactic low dose aspirin to prevent thrombus formation. At present, there is no evidence base to suggest one particular anti-epileptic drug above another, thus use of local clinical guidelines is recommended. Acute rescue treatment of seizures with benzodiazepines or if ineffective, intravenous phenytoin or phenobarbitone is recommended in India (Sarah, 2007). PWS may be treated with cosmetic camouflage creams, pulsed tunable dye laser and cosmetic surgery. Laser therapy is the most effective approach to therapy of the PWS, but results are extremely variable being considered satisfactory in only 45% cases. The development of a multicentre network for SWS trials has been proposed due to current controversies in management of the seizures, other neurologic impairment and the skin lesions. Early surgery is advocated for better seizure control and to prevent developmental delay (Rochkind, 1990).

The differential diagnosis of SWS includes other varieties of arteriovenous malformation as Klippel Trenaunay Weber syndrome, Rendu-Osler Weber syndrome, Bannayan Riley Ruvalcaba syndrome, Divry Van Bogart syndrome and Cobb syndrome. However exact categorisation of the lesions is not always possible due to overlapping features in many of these syndromes (Arif et al., 2008).
Patient dental education and implementation of preventive procedures help in avoiding gingivectomy and endodontic procedures which can be risky to these patients. Treating these patients can be challenging to the general dental practitioner and when a surgical procedure is planned, achieving haemostasis can be a significant problem. Practitioners should be prepared in case any complication ensues and have a suitable armamentarium at their disposal for haemostasis. Over instrumentation during endodontic procedures should be prevented to avoid haemorrhage in periapical regions. Behavioural problems may be encountered either due to previous exposure to hospital settings or due to mental impairment which demands the use of behavioural management techniques.

Conclusion

Diagnosis and treatment of SWS relies on a detailed history, thorough clinical examination and imaging requiring a multidisciplinary approach and co-ordination between different fields of medicine, surgery and dentistry. The importance of newborn diagnosis and further multicentre clinical investigation as well as continuing genetic research is essential in SWS. Determining the underlying cause of SWS is a difficult problem but further research in the area would allow an increase in understanding and application of new treatment options.

Acknowledgements

We would like to thank our Professor and Principal Dr. H. N. Shama Rao for his support and guidance.

References

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