Case Series

STURGE WEBER SYNDROME

Ghulam Ali Memon, Abdul Razzaque Memon and Imdad A. Khushk

ABSTRACT

Six cases of Sturge Weber Syndrome are reported which presented at Plastic Surgery ward of Liaquat University Hospital Jamshoro, Pakistan. It includes characteristics of patients, management of cases and their outcome. Male female ratio was same in these cases and their age was above 30 years. Outcome of the management of this syndrome was found dependent upon case to case. As Sturge Weber Syndrome is a very rare congenital anomaly, hence, its awareness among the plastic surgeons, neurosurgeons and eye surgeons is important.

KEY WORDS: *Sturge Weber Syndrome. Port wine stain. Eye problems. Pakistan.*

INTRODUCTION

Sturge Weber syndrome (SWS) is a neurocutaneous disorder classically presenting with a facial port-wine stain, vascular eye abnormalities, and an ipsilateral occipital leptomeningeal angioma[1]. In SWS, the port wine stain is noted at birth and generally occurs on the same side as the excessive blood vessel growth (leptomeningeal angiomatosis) in the brain is accompanied by the accumulation of calcium (intracranial calcifications). The port wine stain primarily occurs along the distribution of the trigeminal nerve on the face, though in some cases, it may not appear at all. Although the discolouration usually affects only one side of the face, a slight extension over the midface occurs in fifty per cent of the cases. The port wine stain tends to deepen in colour with age, and nodular elevations may also develop. Port wine stains on lips and mucous membrane lining of the mouth are present in approximately 25 per cent of the patients [2].

Seizures occur in more than half of the patients, usually beginning during the first year of life [3]. These tend to become more frequent and severe with age. A form of paralysis (hemiparesis or hemiplegia) occurs in 30 per cent of patients. Mental disturbances occur in fifty to sixty per cent of patients.

Seventy per cent of patients have associated eye problems on the same side of the face as the port wine stain and clumps of blood vessels (leptomeningeal angiomatosis) with intracranial calcifications. The eye problems do not tend to occur in SWS patients who have no port wine stains. In many cases, the eye lesion is a glaucoma, and typically presents at birth and accompanied by the enlargement of the eyeball (buphthalmos), but it may begin anytime before or after the age of two years[4].

The exact cause of SWS is not known. In some, it is believed to be an autosomal dominant hereditary disorder. But, it may also be caused by trauma or viral infection sustained during early gestation.

PRESENTATION OF CASES

Six cases of SWS included 3 males and 3 females. All the cases reported during 2000-2003 and were above the age of 30 years. Five cases presented with oro-facial port wine stain; angiomatosis alongwith cutaneous and mucous lesions localised in the area of distribution of the first and second branches of trigeminal nerve in association with right or left upper and lower lip and the cheek hypertrophy. All cases had drooped cheeks and lower lips. Of these five cases, one male patient presented with secondary glaucoma resulting in blindness of one eye. One case presented with forehead angiomatosis and history of seizures. Patients with blindness and seizures were treated conservatively. Surgical treatment was offered in four cases. In three patients; two females and one male, with excessive drooping of the lower lip and cheek, excessive tissues of the lower lip along with the cheek were excised and the defects closed by primary reconstruction. One female patient presenting with nodular papillomatosis of cheek was treated by 95 per cent excision of the lesion and reconstruction of the cheek was done by advancement of a rotation flap **(Figures I and II)**.

Four patients operated in this series presented with hypertrophy of the upper and lower lips and cheek

leading to facial asymmetry and dento-skeletal malocclusion. At surgery, the excessive tissue was excised and the defect was closed primarily in three cases while in one patient reconstruction of the cheek had to be carried out by the advancement of a rotation flap. At follow-up, all patients had improved looks with resultant elation of mood and enhanced self-confidence. Reduction in the drooping of the cheek and lips resulted in a competent oral sphincter and complete cessation of the dribbling of the saliva. It also theoretically prevented the development of future complications like mucosal ulceration, infection and haemorrhage.

One patient in this series presented with associated seizures which was treated conservatively by the neurosurgeon with good response.

Figure I: Facial appearance of a female patient with SWS before surgery

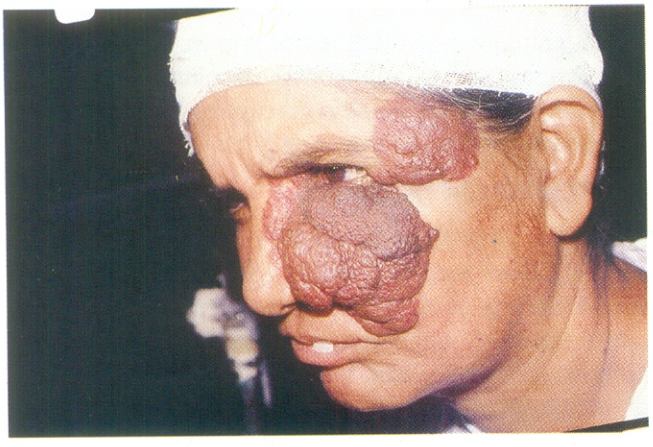
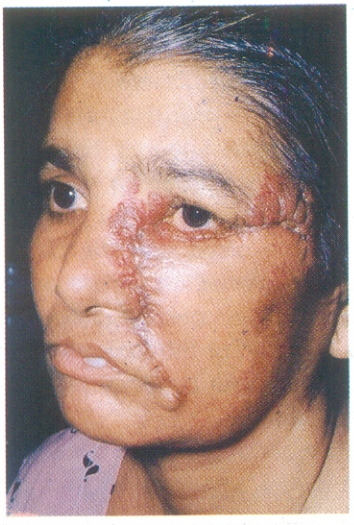


Figure II: Facial appearance of female patient with SWS postoperatively



DISCUSSION

Clinically, the full-blown condition of SWS consists of a facial port-wine stain (PWS) involving the V_1 facial trigeminal skin area, alone or in combination with V_2 and V_3PWS, seizures and ocular abnormalities (glaucoma and choroidal angioma). Radiologically, a leptomeningeal (pial) capillary and venous malformation, mostly located in the parieto-occipital area, cerebral atrophy and calcifications are demonstrated[5]. Glaucoma is present in approximately half of the cases. In our series of six patients, four patients presented with muco-cutaneous lesions with no associated ocular problems or seizures. Of the remaining two cases, one patient presented with glaucoma leading to blindness and one presented with seizures.

The advent of the laser therapy has dramatically improved the outcome in patients with port wine stain. However, patients with SWS present with bulky haemangiomas of the cheeks and lips which require debulking surgery to achieve cosmetic and functional improvement.

Associated glaucoma has been widely reported to be present in 50 per cent of the patients with SWS. This is quite contrast to our series where only one patient (16.7%) presented with eye problem. This can be related to the small number of cases in this study. Patients with SWS often present with seizures during the first year of life. Currently, only patients with clinically significant seizures who do not respond to medical treatment are candidates for early epileptic surgery[6]. The only case in this series with associated seizures, however, presented late and responded to conservative treatment. In conclusion, SWS is a very rare congenital anomaly, hence, its awareness among the plastic surgeons, neurosurgeons and eye surgeons is important.

REFERENCES

1. Comi AM. Pathophysiology of Sturge-Weber syndrome. J Child Neurol. 2003;18(8):509-16
2. Gasparini G, Perugini M, Vetrano S et al. Angiodysplasia with osteohypertrohy affecting the oromaxillofacial area: clinical findings. J Craniofac Surg 2001; 12(5): 485-9
3. Saneto RP, Wyellie E. Epilepsy surgery in infancy. Semin Pediatr Neurol 2000; 7(3): 187-93
4. Amirika A, Scott IU, Murray TG. Bilateral diffuse haemangiomas with unilateral facial nevus flammeus in Sturge-Weber syndrome. Am J Ophthalmol 2000; 130(3): 362-4

5. Boukobza M, Enjolras O, Cambra M et al. Sturge-Weber syndrome. The current neuroradiologic data [Article in French] J Radiol. 2000 ;81(7):765-71.

6. Kramer U, Kahana E, Shorer Z et al. Outcome of infants with unilateral Sturge-Weber syndrome and early onset seizures. Dev Med Child Neurol 2000; 42(11): 756-9

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