ORIGINAL ARTICLE

Sturge-Weber Syndrome – Case Series

Dr. Ghulam Ali Memon

Liaquat University of Medical & Health Sciences, Jamshoro

SUMMARY. Objective: The author reports six cases of Sturge-Weber Syndrome.

Setting: Plastic Surgery Ward 18, Liaquat University Hospital, Jamshoro.

Method: Descriptive presentation of six cases of Sturge-Weber Syndrome characteristics of the patients, surgical procedures and outcome noted.

Results: Results of this syndrome are usually dependent upon case to case.

Conclusion: This is a very rare congenital anomaly and hence its awareness to the plastic surgeons, neurosurgeons and eye surgeons is important.

Key Words: Sturge-Weber Syndrome, port wine stain, eye problems.

Nevus flammeus is the discoloration of the face which mimics the red colour of port wine. In Sturge-Weber Syndrome (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 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 midline 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[1].

Seizures occur in more than half of the patients, usually beginning during the first year of life[2]. These tend to become more frequent and severe with age. A form of paralysis (hemiparesis or hemiplegia) occurs in 30% 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 Sturge-Weber patients who have no port wine stains. In many cases, the eye lesion is a glaucoma, typically present at birth and accompanied by the enlargement of the eyeball (buphthalmos), but it may begin anytime before or after the age of two years[3].

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

Patients & Methods

Six cases of Sturge-Weber Syndrome, three males and three females are presented. All the cases were above the age of 30 years. Five cases presented with oro-facial port wine stain angiomatosis with cutaneous and mucous lesions localised in the area of distribution of the first and second branch of trigeminal nerve in association with right upper and lower lip and cheek hypertrophy.

Professor Dr. Ghulam Ali Memon *DSS, FCPS*
Professor of Surgery and Plastic Surgery
Dean Faculty of Surgery and Allied Sciences
Liaquat University of Medical and Health Sciences, Jamshoro
Correspondence to author
Paper received 11th January 2004
Accepted 28th January 2004.

All cases had drooped cheeks and lower lips. Of these five cases, one male patient presented with secondary glaucoma ending 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% excision of the lesion and reconstruction of the cheek by advancement of a rotation flap.

Case 1: (Fig. 1 & 2) Female with lower lip hypertrophy, right cheek and right neck port wine stain. Treated with lip reduction.

Case 2: (Fig. 3) Female with lower lip hypertrophy and left cheek port wine stain. Treated by lip reduction.

Case 3: (Fig. 4) Male with lower lip hypertrophy, right cheek, neck and upper chest port wine stain. Treated by lip reduction.

Case 4: (Fig. 5 & 6) Female with nodular papillomatosis of the left lower lid, infra-orbital area and left temple. Treated by excision and cheek rotation flap.

Case 5: (Fig. 7 & 8) Male with upper lip hypertrophy and port wine stain of the left cheek. Associated glaucoma causing blindness of the left eye.

Case 6: (Fig. 9) Male with left forehead angiomatosis and history of seizures. Treated medically for epilepsy.

Results

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.

Discussion

The Sturge-Weber syndrome consists of a unilateral port-wine haemangioma of the skin along the trigeminal distribution and is accompanied by an ipsilateral leptomeningeal angioma causing seizures. Glaucoma is present in approximately half of the cases[4]. In our series of six patients, four patients presented with mucocutaneous 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 pulse dye laser therapy has dramatically improved the outcome in patients with port wine stain. However, patients with Sturge-Weber syndrome 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% of the patients with Sturge-Weber syndrome. This is quite contrast to our series where only one patient presented with eye problem. This can be related to the small number of cases in this study.

Patients with Sturge-Weber syndrome 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[5]. The only case in this series with associated seizures, however, presented late and responded to conservative treatment.

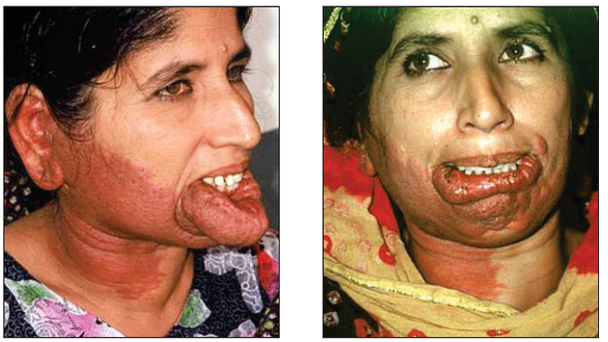

Fig 1

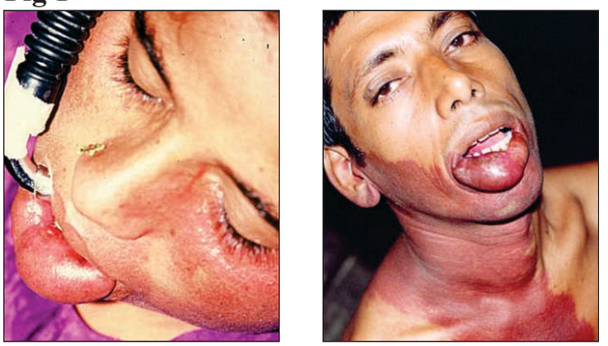

Fig 2

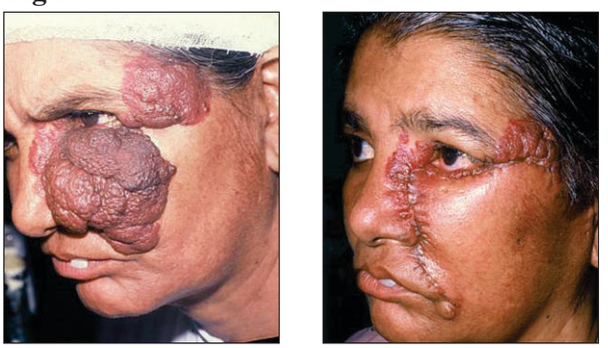

Fig 3

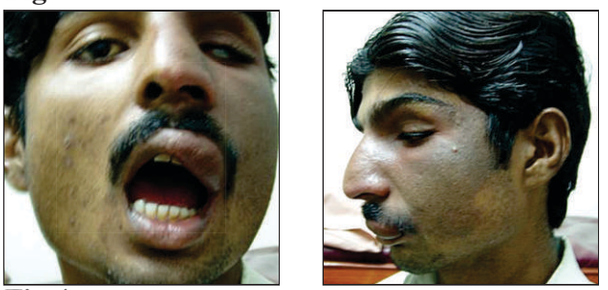

Fig 4

References

1. Gasparini G, Perugini M, Vetrano S, Cassoni A, Fini G. Angiodysplasia with osteohypertrohy affecting the oromaxillofacial area: clinical findings. J Craniofac Surg. 2001; 12(5): 485-489.
2. Saneto RP, Wyellie E. Epilepsy surgery in infancy. Semin Pediatr Neurol. 2000; 7(3): 187-193
3. Amirika A, Scott IU, Murray TG. Bilateral diffuse haemangiomas with unilateral facial nevus flammeus in Sturge-Weber syndrome. Am J Ophthalmol. 2000; 130(3): 362-364.
4. Lee H, Choi SS, Kim SS, Hong YJ. A case of glaucoma associated with Sturge-Weber syndrome and Nevus of Ota. Korean J Ophthalmol. 2001; 15(1): 48-53.
5. Kramer U, Kahana E, Shorer Z, Ben-Zeev B. Outcome of infants with unilateral Sturge-Weber syndrome and early onset seizures. Dev Med Child Neurol. 2000; 42(11): 756.

★ ★ ★ ★ ★