

Case report

Bilateral Sturge Weber Syndrome- a rare case report

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Abstract

Background: Sturge-Weber syndrome is a rare congenital neuro- oculo- cutaneous disorder. **Objective:** To report a very rare unusual case of bilateral manifestation of Sturge Weber syndrome. **Case:** We report an unusual case of a 17-year-old female with advanced stage of bilateral glaucoma associated with facial nevus extending to the other half of the face as well and bilateral intracranial calcification. **Conclusion:** Sturge -Weber syndrome can manifest as a bilateral condition.

Keywords: facial nevus, leptomeningeal angioma, glaucoma

Introduction

Sturge-Weber syndrome (SWS), also called encephalo-trigeminal angiomatosis is a sporadic neuro-oculo-cutaneous syndrome characterized by the presence of facial nevus, ocular abnormalities (glaucoma, choroidal hemangioma) and leptomeningeal angioma mostly involving occipital and posterior parietal lobes (Baselga et al, 2004). Sturge -Weber syndrome is a rare disorder that occurs with a frequency of approximately 1 per 50,000.

The cause of the disease is believed to be the persistence of vascular plexus around the cephalic portion of the neural tube. This plexus develops during the sixth week of intrauterine development but normally undergoes regression during the ninth week. The common clinical manifestations of SWS include progressive seizures, unilateral cutaneous vascular nevus following the ophthalmic divisions of the trigeminal nerve, ipsilateral glaucoma, contralateral hemiparesis, hemiatrophy, hemianopia

and mental retardation. These changes are usually unilateral and can be seen equally in both sexes with no racial differences (Govori et al, 2010). The radiographic hallmark of SWS is “tram-line” or gyriform calcifications usually involving the occipital and parietal lobe.

Case report

A 17-year-old female presented with progressive, painless diminution of vision in both the eyes. Her vision in the right eye was PL with no accurate PR and that in the left was hand movement close to her face. The patient was the only child of a non-consanguineous marriage and her birth had been at full term by normal delivery. The family history did not reveal any similar complaints from her immediate or distant relatives. The patient had a history of convulsions. The last episode had taken place two years ago. She had not taken any medicines for this disorder. No evidence of mental retardation was noted.

Physical examination revealed a port wine nevus on the right side of face extending towards the left side as well and her father reported that this had been present since birth (Figure 1).

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Consent

An informed consent for publication of this case report and accompanying images was obtained from the patient's father and the patient as well.



Figure 1: Facial naevus on both side

Intraoral examination revealed hyper-vascularity of the right sided palate. The palate in the left side appeared normal. A unilateral vascular involvement of the soft palate and buccal mucosa was noticed. A hemangioma in the upper lip was also noted (Figure 2).

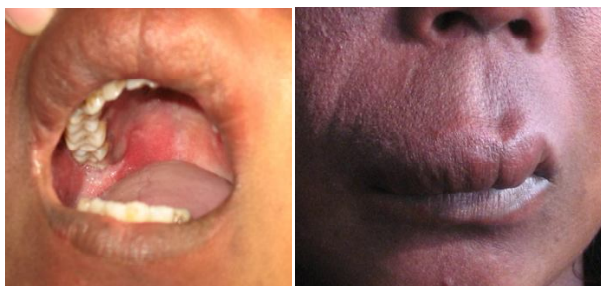


Figure 2: Hypervascularity of right palate and haemangioma of upper lip

Ophthalmic examination showed a relative afferent pupillary defect in the right eye and advanced glaucomatous cupping in both the eyes (Figure 3). No choroidal hemangioma was observed. The intraocular pressure (IOP) was elevated in both the eyes. The patient was started on B blocker eye drop two times a day in each eye. The patient responded well to the topical B blocker in the subsequent follow-up when the IOP was found to be well controlled. The counseling for poor visual prognosis was done since she presented with advanced glaucomatous optic atrophy.

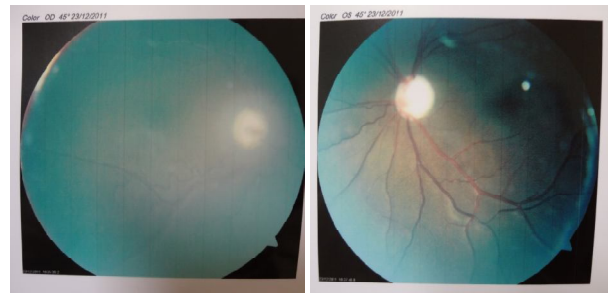


Figure 3: BE glaucomatous optic atrophy

The CT scan of the head showed intracranial calcification in the occipital lobe extending superiorly to the right parietal lobe (Figure 4). The cerebellar hemisphere was spared.

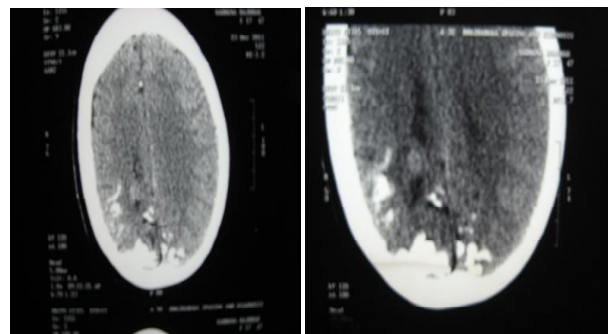


Figure 4: Occipital calcification extending superiorly to the right parietal lobe

Discussion

Sturge-Weber syndrome was first described by Schirmer in 1860 and later more specifically by Sturge in 1879, who associated dermatological and ophthalmic changes of the disease to neurologic manifestations. Weber in 1929 complemented it with the documentation of radiologic alterations seen in these patients. Sturge-Weber is

an embryonal developmental anomaly resulting from errors in mesodermal and ectodermal development. Unlike other neurocutaneous disorders (phakomatoses), Sturge-Weber occurs sporadically (Moe et al, 2003).

Sturge-Weber syndrome is characterised by neurological problems and a birthmark, known as a facial nevus or a port-wine stain. Port wine stains are classically faint, pink macules, tend to darken



progressively to red purple, may be isolated with a well-delineated border or may be very diffuse. It has usually unilateral distribution along one or more segments of the trigeminal nerve. Occasionally, bilateral involvement or additional port wine lesions are found elsewhere on the body (Neville et al, 2002). According to INAN (1999), the port wine nevus is localized in the face, especially on the right side, and is detected in 87% to 90% of the cases. The lesion extension over the middle line is observed in 50% of the patients and bilateral involvement can be detected in about 33% of the cases. In the present study, bilateral involvement of the face was seen. The lesion was extensive on the right side extending towards the left side of the face as well.

Oral changes occur in 40% of cases of this syndrome and may include massive growth of the gingival tissues, hypervascular changes in the palate, haemangioma of the lip and asymmetric jaw growth (Wilson S et al, 1986). In this case report, the patient had a massive hemangioma on the right upper lip and a hypervascular palate as well.

Affected individuals also have leptomeningeal angioma that overlie the ipsilateral cerebral cortex. Meningeal angiomatosis is usually associated with a convulsive disorder and sometimes contralateral hemiplegia and mental retardation (Neville et al, 2002). In the present study, the patient had a history of convulsions. She had not taken any medications for this disorder. Convulsions occur in approximately 75% of patients, and 75% of the seizures appear within the first year of life. The majority of seizures are partial motor or complex partial type.

Glaucoma is the most common serious eye problem of SWS, with a reported incidence of 30-70% (Govori et al, 2009). Glaucoma may be present at birth or develop later. Pressure within the eye may damage the optic nerve, usually in the eye on the same side as the birthmark. The reason for this increased eye pressure may be the result of the outflow obstruction by a vascular malformation of the front area of the eye (Govori et al, 2009). In the present study, the patient had glaucomatous cupping

in both eyes.

Bilateral intracranial involvement, reported in about 15 % of cases, is associated with an earlier onset of seizures and worse cognitive development compared to unilateral cases (Bebin et al, 1988). Our patient had bilateral intracranial involvement. Calcification in the damaged cortical layer may become visible on X-ray of the skull and CT scan of the brain, and is often curvilinear with a railroad track pattern ipsilateral to the port-wine stain. The usual sites of calcification are the occipital and posterior parietal lobes. But it can also affect other cortical regions and both cerebral hemispheres (Kumar et al, 2004).

In the present case, the CT scan of the brain revealed a calcification in the occipital lobe extending superiorly to the right parietal lobe.

SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other (Roach et al, 1992). Our patient had complete Sturge - Weber syndrome.

The diagnosis of Sturge-Weber syndrome can frequently be suspected when a patient is noted to have a facial port-wine birthmark associated with ipsilateral either brain or eye involvement. Our case is interesting due to presence of bilateral facial naevus, bilateral glaucoma and bilateral intracranial calcification.

Conclusion

Sturge-Weber syndrome is a rare phacomatosis which may present as a bilateral condition.

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