CASE REPORT

Sturge-Weber Syndrome in a 56year Old Woman: A Case Report

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ABSTRACT

Background: *Sturge-Weber syndrome is a sporadic phacomatoses with angiomas involving the leptomeninges and skin of the face, typically in the ophthalmic and maxillary distributions of the trigeminal nerve. Glaucoma is a common ocular manifestation. Presentation is typically at birth with a facial angioma.*

Method: *Case note of a patient with a diagnosis of Sturge-Weber syndrome was used and the relevant literature reviewed.*

Result: *A 56 year old woman with pain, photophobia in the left eye of 6 months duration. She had lost vision in the eye about 20 years earlier and was born with a dark patch on the left side of her face. She had no previous history of convulsion. On examination, she had a port-wine stain involving the left side of her face. Examination of the left eye revealed a visual acuity of no light perception with episcleral haemangioma. There was a relative afferent pupillary defect and fundoscopy revealed a pale pathologically cupped disc with tortuous retinal vessels. The intraocular pressure was elevated.*

Conclusion: *Sturge-Weber syndrome is a rare phacomatoses which may present with ocular complications such as glaucoma. If glaucoma is left untreated decreased vision and blindness result. People of any age therefore with port-wine stain in the ophthalmic distribution of the trigeminal nerve should have yearly eye examination and measurement of intraocular pressure, regardless of whether they have symptoms or not.*

KEYWORDS: *Sturge-Weber syndrome; encephalotrigeminal angiomatosis; port-wine stain.*

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INTRODUCTION

Sturge-Weber syndrome (SWS) also known as encephalotrigeminal angiomatosis is a sporadic phacomatosis which presents at birth [1]. Its exact incidence is not known[2]. Males and females are equally affected [3] and no racial predilection is known[2]. It is characterized by cutaneous facial angioma, leptomeningeal angioma associated with seizures and other neurologic complications and glaucoma [4]. The cutaneous angioma is called a port-wine stain(PWS).

Neurologic manifestation vary depending on the location of the leptomeningeal angiomas which most commonly are located in the occipital and parietal region and the secondary effects of the angioma[5].

Glaucoma may occur in 30-71% of patients with SWS and treatment should be aimed at controlling the intraocular pressure and preventing progressive visual loss and blindness[5]. We report a case of Sturge-Weber syndrome diagnosed in a 56year old woman.

CASE REPORT

A 56 year old woman presented at the eye clinic, Jos University Teaching Hospital on April 27, 2004 with a 6 months history of pain, tearing and photophobia in her left eye. There was no eye discharge or any history of preceding trauma to the eye. She had lost vision in the eye about 20 yrs earlier and was born with a dark patch on the left side of her face. There was occasional headache associated with the eye pain. She had used chloramphenicol eye drop in the past without any relief. There was no previous history of seizures, loss of consciousness or weakness of any part of her body.

Examination revealed a port-wine stain (PWS) on the left side of her face involving the forehead, upper and lower eyelids, cheek and upper lip. She was conscious and well oriented in time, place and person. She was of normal intelligence.

On ocular examination, the right eye had a visual acuity (VA) of $^{6}/_{9}$ with a normal anterior segment. Findings on fundoscopy were within normal limits with a cup disc ratio (CDR) of 0.3. Intraocular pressure (IOP) measured 12mmHg by applanation tonometry at 9:30a.m. Visual field examination did not reveal a hemianopia.

The left eye had a VA of no light perception with episcleral haemangiomas inferonasally. There was corneal epithelial oedema. The cornea was however not enlarged. The pupil was 3mm, round with a relative afferent pupillary defect. Dilated fundoscopy revealed a pale pathologically cupped disc (CDR 1.0) indicating chronic glaucoma with tortuous retinal vessels. The IOP was 42 mmHg at 9.30a.m.

Based on the findings on clinical examination a diagnosis of Sturge-Weber syndrome was made. She had a skull X-ray which did not reveal any abnormality. She was placed on anti-glaucoma drugs; topical timolol in her left eye and

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acetazolamide tablets 250mg twice daily for 2weeks. At follow-up 2weeks later, the eye pain had resolved and the cornea was clear. The IOP measured 22mmHg at 11.00a.m.

Figure 1. Photograph of the patient.

DISCUSSION

SWS is apparently hamartomatous in nature from persistence of a primitive embryonal vascular plexus. During the sixth week of intrauterine life, this plexus develops around the cephalic portion of the neural tube and under the ectoderm in the region destined to become facial skin. In SWS the vascular plexus fails to regress, as is normal during the ninth week, resulting in angiomatosis of the related tissue[3].

The syndrome may be classified into two; trisystem when it involves the face, leptomeninges and eyes, and bisystem when it involves the face and eyes or the face and leptomeninges [1]. Our patient had a bisystem disorder.

The main clinical features of this syndrome are venous angiomatosis of the leptomeninges of the cerebral cortex, usually unilaterally, ipsilateral facial angiomatosis, ipsilateral gyriform calcification of the cerebral cortex, epileptic convulsions (contralateral focus) or other seizures, ocular defects (choroidal angioma, glaucoma), mental retardation, contralateral hemiplegia, obesity, oral mucosal and gingival involvement [3]. The most striking clinical feature however is the facial naevus (port-wine stain) which generally follows the distribution of one or more divisions of the trigeminal nerve [3]. Not all people with a PWS have SWS; the overall incidence of SWS has been reported to be 8-33% in those with a PWS [5]. For the syndrome to occur in those with a PWS, the area supplied by the ophthalmic division of the trigeminal nerve must be involved [6]. In a patient with dark skin pigmentation a PWS may be difficult to visualize.

Neurologic and developmental morbidity in SWS include seizures, weakness, strokes, headaches, hemianopia, mental retardation and developmental anomalies[5]. Neurologic dysfunction results from secondary effects of the leptomeningeal angiomas on surrounding brain tissue which include hypoxia, ischaemia, venous occlusion, thrombosis, infarction or vasomotor phenomenon[5]. The central nervous system was most likely not affected in the above case as there were no features of neurologic morbidity.

Features of SWS in the ipsilateral eye include glaucoma, conjunctival or episcleral haemangioma, choroidal haemangioma, retinal vascular tortuosity, iris heterochromia, retinal detachment and strabismus [7]. The choroid is the site of the most significant purely vascular anomaly of the eye associated with SWS [8]. In a majority of cases of SWS with ocular involvement, increased number of well- formed choroidal vessels give the fundus a uniform bright red or red orange colour that has been compared to tomato catsup [8].

Glaucoma usually occurs only with an ipsilateral facial PWS, although it may be bilateral when facial involvement is bilateral [5]. Only those with involvement of the ophthalmic division of the trigeminal nerve are at risk of glaucoma[9]. Presentation of glaucoma in 60% of patients is within the first 2 years of life with buphthalmus. The remainder manifest at any time from infancy to adulthood [10]. The absence of buphthalmus in the above case suggests a later onset of the glaucoma.

The pathogenesis of glaucoma is controversial[10]. Isolated trabeculodysgenesis and raised episcleral venous pressure associated with an arteriovenous communication in an episcleral angioma have both been implicated [10]. Decreased vision and blindness as in the above patient, result from untreated glaucoma with intraocular pressure leading to optic nerve damage.

Work up of patients with SWS include neuroimaging studies, EEG and cerebrospinal fluid (CSF) analysis. Besides the clinical examination, neuroimaging studies have been the procedure of choice to establish the diagnosis [5]. The skull X-ray may show the classical "tram-line" or "tram track" or "trolley track" calcification considered pathognomonic for SWS [5]. CT scan may show calcification, brain atrophy, ipsilateral choroid plexus enlargement, abnormal draining veins [5]. MRI is less sensitive than CT for identifying calcification [8] but is

more efficient in the detection of radiological findings related to the clinical status; seizure control, degree of psychomotor development and hemiparesis [11]. It is therefore the imaging modality of choice in these patients[11]. The absence of radiological features of SWS in the above case further confirms that the central nervous system was not involved.

EEG is used for evaluation of seizures and for location of seizure activity when epilepsy surgery is considered [5]. CSF analysis may reveal elevated protein presumably secondary to microhaemorrhage [5].

Treatment of patients with SWS is determined by clinical manifestation. Medical care includes anticonvulsants for seizure control, glaucoma treatment to reduce IOP[5]. Surgical options such as hemispherectomy, corpus capsulotomy are available for seizures refractory to medical treatment[5]. Glaucoma surgery such as goniotomy, trabeculectomy and cyclodestructive procedures may be beneficial if medications are unable to lower IOP [5]. Erbium laser therapy is available for the PWS. If it is carried out during early life it is effective in decreasing the amount of skin discolouration in relatively flat or mildly hypertrophic lesion [12].

In conclusion SWS is a rare phacomatoses which may present with ocular complications such as glaucoma. If glaucoma is left untreated, reduced vision and blindness result. People of any age therefore with PWS in the ophthalmic distribution of the trigeminal nerve should have yearly eye examination and measurement of IOP, regardless of whether they have symptoms or not.

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