CASE REPORT ON STURGE WEBER SYNDROME

Neha Adlakha

1Junior Resident, Department of Ophthalmology, G. R. M. C, Gwalior.

ABSTRACT

The objective of this study is to report a rare case of Sturge Weber Syndrome. Sturge–Weber syndrome, sometimes referred to as encephalo-trigeminal angiomatosis, is a rare congenital neurological and skin disorder. It is one of the phakomatoses and is often associated with port-wine stains of the face, glaucoma, seizures, mental retardation, and ipsilateral leptomeningeal angiomata. A 7 year old male patient presented to OPD with presenting complaints of diminution of vision in right eye since birth. He also complained of hyper pigmented birth mark over right side of cheek and upper part of right side of neck since birth, convulsions and weakness over left side of body since birth. As per the past history, patient had delayed milestones as per age. Complete ophthalmological examination of patient was performed including visual acuity, fundus examination,planation tonometry and measurement of corneal diameter using calipers. Ophthalmological examination showed reduced visual acuity in right eye, glaucomatous optic neuropathy in fundus in right eye, buphthalmos and raised intraocular pressure (24mm) in right eye. Neurological examination revealed reduced tone, reflexes and power in left side of body. Contrast enhanced MRI Brain was performed which showed diffuse cerebral atrophy with dense lepto-meningeal enhancement involving cerebral sulci, sylvian fissure and basal cisterns. 16 channel EEG of patient was normal. Port wine stains were found on right side of cheek and upper part of right side of neck.

KEYWORDS
Encephalotrigeminal, Angiomatosis, Phakomatoses, Leptomeningeal, Milestones, Applanation Tonometry, Glaucomatous, Neuropathy, Buphthalmos, Sylvian.


INTRODUCTION

Sturge Weber Syndrome (SWS) is a dermato-oculo-neural syndrome characterized by cutaneous facial nevus flammeus, ipsilateral cavernous haemangiomata of choroid and ipsilateral meningeal haemangiomatosis. The classical cutaneous feature of SWS is facial nevus flammeus, flat to moderately thick zone of dilated telangiectatic cutaneous capillaries lined by a single layer of endothelial cells in dermis.[1] The lesion is usually unilateral and frequently involves regions of face innervated by first, occasionally first and second and rarely all three branches of trigeminal nerve. The characteristic CNS manifestation of SWS is ipsilateral leptomeningeal haemangiomata which causes atrophy of cortical parenchyma, seizures and mental retardation.[2] CNS lesions are detectable by MRI or CT scan.[3] In many patients, affected meninges become irregularly calcified during life, in which case CNS vascular lesion can be detected on routine skull radiographs. However, patients who have SWS and develop seizures or progressive mental retardation probably need periodic neurological evaluation and intermittent evaluation by CT or MRI brain to rule out treatable lesions or disorders. Gibis et. al report that glaucoma occurs in about one third of patients who have SWS.[4] The glaucoma is usually unilateral on side of nevus flammeus, but bilateral involvement has been reported.

Neurological manifestations include seizures, paralysis and visual field defects from intracranial involvement. A choroidal haemangioma may be present and visible on fundus examination.

Treatment of patients who have SWS is generally symptomatic and directed toward complications caused by vascular lesions of brain and eyes. Seizures are treated medically. Intractable seizures and progressive mental deterioration are sometimes treated surgically by subtotal hemispherectomy.[5] The facial nevus flammeus can be treated by dermatological laser therapy. This treatment frequently results in marked regression of vascular birthmark and substantial cosmetic improvement. Treatment of glaucoma in cases where angle appears similar to primary infantile glaucoma, goniotomy is reasonable first step.

For cases presenting later in life, trabeculectomy, cyclodestruction and glaucoma drainage devices have been advocated.[6,7] In cases where increased episcleral venous pressure is cause of elevated intraocular pressure, a fistulisation procedure (trabeculectomy or drainage implant is more likely to succeed. The presence of choroidal haemangiomas in these patients increases the risk of choroidal expansion or haemorrhage intraoperatively; some surgeons advocate use of prophylactic posterior sclerotomies prior to entry into anterior chamber.

The life expectancy of patients who have SWS appears to be reduced substantially compared with persons in general population.[8] However most early deaths occur in individuals with profound mental retardation and intractable seizures and not with limited form of disease, normal intellectual ability and no seizures.
CASE REPORT

7 year old male patient presented to OPD with presenting complaints of diminution of vision in right eye since birth. He also complained of hyper pigmented birth mark over right side of cheek and upper part of right side of neck since birth, convulsions and weakness over left side of body since birth. As per the past history, patient had delayed milestones as per age.

<table>
<thead>
<tr>
<th>MILESTONE</th>
<th>NORMAL AGE</th>
<th>PATIENT’S AGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neck holding</td>
<td>3 months</td>
<td>11 months</td>
</tr>
<tr>
<td>Rolls over</td>
<td>5 months</td>
<td>1 year</td>
</tr>
<tr>
<td>Monosyllables</td>
<td>6 months</td>
<td>15 months</td>
</tr>
<tr>
<td>Unidextrous reach</td>
<td>6 months</td>
<td>11 months</td>
</tr>
<tr>
<td>Sitting without support</td>
<td>8 months</td>
<td>12 months</td>
</tr>
<tr>
<td>Bisyllables</td>
<td>9 months</td>
<td>16 months</td>
</tr>
<tr>
<td>Stands with support</td>
<td>9 months</td>
<td>15 months</td>
</tr>
<tr>
<td>Walks</td>
<td>15 months</td>
<td>2 years</td>
</tr>
<tr>
<td>Runs</td>
<td>18 months</td>
<td>2.5 years</td>
</tr>
<tr>
<td>2-3 word sentences</td>
<td>2 years</td>
<td>4 years</td>
</tr>
</tbody>
</table>

Clinical Picture

Complete ophthalmological examination of patient was performed including visual acuity, fundus examination, applanation tonometry and measurement of corneal diameter using calipers.

OCULAR EXAMINATION

<table>
<thead>
<tr>
<th>EXAMINATION</th>
<th>RIGHT EYE</th>
<th>LEFT EYE</th>
</tr>
</thead>
<tbody>
<tr>
<td>VISUAL ACUITY</td>
<td>CF 1 FEET</td>
<td>6/6p</td>
</tr>
<tr>
<td>TONOMETRY</td>
<td>24mmHg</td>
<td>16mHg</td>
</tr>
<tr>
<td>FUNDUS</td>
<td>Red glow seen, media clear, disc size/shape/colour/ margin normal, cup/disc 0.9, cup deep, bayoneting sign present, blood vessels normal, general fundus normal, foveal reflex present</td>
<td>Red glow seen, media clear, disc size/shape/colour/ margin normal, blood vessels normal, general fundus normal, foveal reflex present</td>
</tr>
<tr>
<td>CORNEA</td>
<td>13 mm diameter, hazy</td>
<td>9 mm diameter, clear</td>
</tr>
</tbody>
</table>

NEUROLOGICAL EVALUATION

<table>
<thead>
<tr>
<th>TEST</th>
<th>RIGHT SIDE</th>
<th>LEFT SIDE</th>
</tr>
</thead>
<tbody>
<tr>
<td>TONE</td>
<td>NORMAL</td>
<td>REDUCED</td>
</tr>
<tr>
<td>POWER</td>
<td>NORMAL</td>
<td>REDUCED</td>
</tr>
<tr>
<td>REFLEXES</td>
<td>NORMAL</td>
<td>REDUCED</td>
</tr>
</tbody>
</table>

Contrast enhanced MRI Brain was performed which showed diffuse cerebral atrophy with dense leptomeningeal enhancement involving cerebral sulci, sylvian fissure and basal cisterns.

Contrast Enhanced MRI

16 channel EEG of patient was normal

16 Channel EEG
Patient had a history of convulsions for which he was prescribed valproate (35mg/kg/day) and carbamazepine (10mg/kg/day).

Port-Wine Stain

Port wine stains were found on right side of cheek and upper part of right side of neck, for which patient was prescribed emollients and mometasone skin ointment for one month and posted for laser one month later.

DISCUSSION

Sturge Weber Syndrome is rare congenital disease characterized by facial angioma, ipsilateral leptomeningeal haemangioma and ocular manifestations including haemangiomatosa of conjunctiva, episclera and retina. Glaucoma is the most common opthalmic complication of SWS occurring in 30–70% of patients. The distribution of cutaneous and cerebral involvement suggests disturbance early in embryonic development (4–8 weeks gestation) when primitive facial structures overlie future occipital lobes of developing brain. Curvilinear densities paralleling cerebral convolutions to produce so called railroad track sign can be demonstrated by means of CT scan. MRI is less sensitive than CT for identifying calcification but may provide better delineation of abnormalities associated with angiomatous malformation that can confirm diagnosis in young children.

These abnormalities include cerebral volume reduction, prominent deep venous system and enlarged choroid plexus. It typically results in seizures and mental retardation. The classic meningeal haemangioma may be associated with calcification seen on skull X-ray, other subtler findings can be neuroimaged with variety of new techniques. Choroidal haemangiomas and episcleral haemangiomas are commonly seen and leakage from choroidal haemangioma may cause retinal oedema. Such ocular abnormalities can be seen with indocyanine green angiography. If glaucoma occurs in infancy, an isolated trabeculodysgenesis type of angle anomaly usually is assumed, described in one case due to abnormalities in canal of Schlemm and juxtacanalicular tissue. Weiss described two mechanisms more common of which occurs in infants, with developmental anomaly of anterior chamber angle.

One histopathologic report described partial developmental anomaly of the anterior chamber angle, and another study revealed neovascularization in trabecular meshwork. Cibis and associates found aging changes similar to seen in chronic open angle glaucoma in trabecular meshwork of three eyes with Sturge Weber Syndrome.

Onset of glaucoma can be at birth or later in childhood. If IOP is elevated during early infancy, enlargement of cornea can occur. When SWS is first suspected, a complete ophthalmic evaluation is essential including measurement of IOP.

MANAGEMENT

Treatment for Sturge Weber syndrome is symptomatic. Laser treatment lighten or remove port wine birthmarks in children. Anticonvulsants are used to control seizures. Physical therapy should be considered for infants and children with muscle weakness. Educational therapy is often prescribed for those with mental retardation or developmental delays. Neurosurgery involving removing portion of brain that is affected by disorder can be successful in controlling seizures so that patient has only few seizures that are much less intense than before surgery. Surgeons may also opt to “switch-off” the affected side of brain.

SWS glaucoma is difficult to treat and there is no universally accepted treatment. Initial therapy with topical drops can be effective when onset occurs late. Surgery is indicated in early onset cases and when medical treatment is inadequate. Adequate long-term pressure control can frequently be achieved, although multiple surgeries are typically necessary. Aqueous shunts have shown promise in management of intractable glaucoma in SWS. Some claim that this is sometimes responsive to goniotosomy with more than one procedure frequently required.

In most cases associated with elevated episcleral venous pressure, medical therapy has limited efficacy and this appears to be true for prostaglandins. In one report, use of latanoprost was associated with development of anterior uveal effusion. Success has been reported with trabeculectomy in children and adults. Other authors prefer trabeculotomies combined trabeculectomy/trabeculotomy or glueca tubes such as an Ahmed valve or a two-staged Baerveldt procedure. As child ages, elevated IOP is due to elevation of episcleral venous pressure that occurs due to arteriovenous shunts through episcleral haemangiomas. In face of surgical failure, cyclodestructive procedures cryotherapy, diode laser cyclophotocoagulation or Endocyclo-Photocoagulation can be performed, although more than one intervention is usually required, as is the case in many such difficult paediatric glaucomas.

A particular risk of glaucoma filtering surgery in SWS is massive choroidal effusion or expansive haemorrhage which is increased in these patients. In one study of 30 patients, goniotosomy was not associated with intraoperative choroidal effusion and expansive haemorrhage and was investigator’s first choice in most cases. Careful attention to maintain normal to high IOP during surgery through use of anterior chamber maintainer cannula for constant infusion, generous amounts of viscoelastic and meticulous wound closure may forestall intra and perioperative complications. Special care must be taken with implanted drainage devices to prevent excessive early postoperative hyptonus.

Choroidal or subretinal fluid accumulation after surgery may be dramatic, but spontaneous resorption occurs within 1–2 weeks. Angle surgery in form of goniotosomy and trabeculotomy has been used successfully in some SWS patients. A few reports exist on effect of Non-penetrating glaucoma surgery on SWS.
Some surgeons prefer to perform one or more prophylactic posterior sclerotomies just before filtration surgery or any other intraocular procedure to reduce risk for choroidal or retinal effusion and expulsive haemorrhage. Another surgical approach to reduce pressure in these patients, while minimizing intraocular complications is implantation of valved or nonvalved glaucoma drainage device. One case series reported good outcomes after two patients, while minimizing intraocular complications is surgery or any other intraocular procedure to reduce risk for prophylactic posterior sclerotomies.

In this situation, the Baerveldt implant is placed in the appropriate location; the tube is reflected to an adjacent subconjunctival location and anchored to sclera. Six weeks later, the tube is dissected free and inserted into anterior chamber.

REFERENCES