Original Article ISSN (O):2395-2822; ISSN (P):2395-2814

Analysis of Sturge Weber Syndrome: A Retrospective Study.

Ruchi Shukla[1], Pankaj Baranwal[1], A. S. Thakur[1], Garima Rai[1]
[1]Department of Ophthalmology, Heritage Institute of Medical Sciences, Varanasi, Uttar Pradesh, India.

Received: August 2018
Accepted: August 2018

ABSTRACT

Background: To review the clinical manifestations and neuroimaging features of patients with Sturge-Weber syndrome (SWS) treated at a tertiary care centre over a 3 year period. **Methods:** A retrospective study of six patients with SWS (4 males and 2 females) was conducted. Data was collected by reviewing the clinical histories of patients diagnosed with SWS over the last 3 years. **Results:** All patients had port-wine stain (PWS) involving the eyelid. Glaucoma was the main ocular disease being diagnosed in 2 eyes of three patients (66.66%). Five patients (83.33%) had neurological impairment including seizure, hemiparesis, headache, and delayed development. However, the most common neurological manifestation was epilepsy (83.33%), which could be controlled with antiepileptic drugs. In neurological imaging intracranial abnormalities were demonstrated which included cerebral atrophy (75.0%), cerebral calcification (50.0%), leptomeningeal angioma (25.0%), and enlarged choroidal plexus (25.0%). The ocular complications and intracranial abnormalities were usually ipsilateral to the PWS.. **Conclusion:** Port-wine stains, glaucoma, and seizure were the most common clinical presentations of Sturge-Weber syndrome detected in this study. Complete ophthalmic and neurological evaluation should be performed at the time of diagnosis.

Keywords: Sturge-Weber, Port-wine stains, Nevus flammeus, Leptomeningeal angiomatosis, Epilepsy.

INTRODUCTION

Sturge Weber syndrome (SWS) is a congenital vascular disease characterized by a facial capillary malformation that is port-wine stain (PWS) associated with venous and capillary malformations in the brain and eyes. Association at other locations, such as the buccal cavity or the respiratory tract, can also be observed.[1] This congenital disease is mainly sporadic rather than hereditary, although some familial cases have been described.[2] Its incidence is between 1/50 000 and 1/230 000 live births.[3]

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.[4] The lesion is frequenly unilateral and commonly involves regions of face innervated by first branch of trigeminal nerve & it may involve lips, gingival, buccal mucosa, palate, and floor of the mouth. The characteristic CNS presentation of SWS is ipsilateral leptomeningeal haemangioma which causes atrophy of cortical parenchyma, seizures and mental retardation.[5]

Name & Address of Corresponding Author
Dr Pankaj Baranwal,
Department of Ophthalmology,
Heritage Institute Of Medical Sciences,
Varanasi, Uttar Pradesh,
India-221311.

Glaucoma is most common ocular manifestation with an incidence of 30%-71% in patients in Sturge Weber syndrome,[6] and It is also associated with vascular malformations of conjunctiva, episclera, choroid and retina. However, the clinical manifestations of SWS are varied widely, with absent or varying neurological and/or ocular features in some cases.[7]

MATERIALS AND METHODS

We conducted a retrospective study by reviewing the data of patients diagnosed with SWS between 2015 and 2018 in the ophthalmology department at a tertiary hospital. Data were collected by reviewing the clinical histories of patients diagnosed with SWS over the last 3 years

Data obtained from each patient's clinical history were as follows:-

- Epidemiological variables: sex and age at presentation.
- Variables related to skin lesions: presence or absence of facial angioma, side affected by the lesion.
- Variables related to ophthalmological symptoms: presence or absence of eye involvement, type of eye disorder, and treatment used.
- Variables related to intracranial lesions: presence of leptomeningeal angiomatosis, extension and location of the lesion, affected side

- Variables related to neurological symptoms: presence or absence of epilepsy, presence or absence of hemiparesis, associated mental retardation, recurrent headache attacks
- Variables related to tests and the results obtained: neuroimaging studies (computed tomography and/or brain magnetic resonance imaging).

RESULTS

We included 6 patients (66.6% males and 33.3% females) diagnosed with SWS at our hospital between 2015 and 2018.

Age at diagnosis of SWS ranged from six months to 50 years, with a mean age of 13 years.

All patients had Port Wine Stain on their face with neurological and/or ocular complications.

Five patients (83.33%) had neurological impairment and three patients (50%) had ocular problem.

All patients had unilateral PWS affecting the first and second sensory branch of trigeminal nerve. Of the patients with facial angioma, 5 displayed right facial angioma (83.33%), 1 on the left side (16.66%).

The ocular complications were ipsilateral to the PWS.

Glaucoma was the main ocular disease observed in two eyes of three patients (66.6%) who presented with ocular problems. Both cases were initially treated with antiglaucoma topical medications, one patient later on needed trabeculectomy for further intraocular pressure control.

Dilated episcleral vessels were demonstrated in two patients (66.6%).

Five patients (83.33%) had neurological impairment. However, the most common neurological manifestation detected was epilepsy (83.33%). Four of five patients (80.00%) had their first seizure during their infancy.

The other neurological findings included hemiparesis (33.33%), delayed development (33.33%), mental retardation (33.33%), headache (25%).

All patients who suffered from seizures could be treated with antiepileptic medications.

Neurological imaging was performed in the all cases (6 patients). Intracranial abnormalities were demonstrated including cerebral atrophy (75.0%), cerebral calcification (50.00%), leptomeningial angioma (25.0%), and enlarged choroidal plexus (25.0%). All of detected leptomeningial angioma involved the occipital lobes and posterior parietal lobe. The intracranial abnormality was also usually ipsilateral to the PWS.

DISCUSSION

SWS was first mentioned in 1860 by Schirmer, a German ophthalmologist, but it was not fully described until 1879 by Sturge. Forty-three years later, in 1922, Weber completed the list of signs by describing the radiological findings.[8]

The manifestations of this syndrome are multiple; one of its salient features is the port-wine stain or facial angiomatosis, which is the most frequent vascular malformation. It manifests in 0.3% of newborns.

Facial angiomatosis is typically located on the eyelid and the forehead, especially in the distribution of the first and second branch of the trigeminal nerve.

Among patients who present with facial PWS, only 8 to 20% develop neurological symptoms however,[9] in our study 66.66% patients presented with neurological impairment.

The risk of associated neurological and/or ocular disorder in a patient with partial or full V1 (forehead and/or eyelid) involvement was 18 to 65% in previous studies, glaucoma and epilepsy being the most common manifestations.[10]

All of our patients had PWS in V1 area, accompany with neurological problem and/or ocular manifestation. However, seizure and glaucoma were the most Common neurological and ocular disorder detected in this retrospective study In our study 83.33% patients presented with neurological impairment and 50% patients manifested glaucoma.

Ocular involvements in SWS arise primarily from vascular abnormalities of the conjunctiva, episclera, retina, and choroid. Ocular manifestations of SWS were episcleral/conjunctival hemangioma, iris heterochromia, glaucoma, choroidal hemangioma, optic disc coloboma, and cataract.[11] The ocular complication is usually ipsilateral to the PWS.

Glaucoma is the most common manifestation, an incidence of 60 to 74.5% has been previously reported in studies and this occurred in 66.66% of our patients who presented with ocular problems.[12,13]

The onset of glaucoma is bimodal. From a literature review, about 60% of patients develop glaucoma in infancy when the eye is susceptible to increased intraocular pressure and 40% in childhood or later.[14] Out of the 2 patients who manifested glaucoma one developed at the age of 8 yrs while another patient presented at 50 years.

Glaucoma is the most common cause of visual loss in patients with Sturge-Weber syndrome.[15] Various mechanisms have been proposed in the past with trabeculo-dysgenesis being responsible in the early onset glaucoma and increased episcleral venous pressure contributing to the raised intraocular pressure later on.[16]

The onset of glaucoma at birth or during infancy is a common feature.[15] This was also evident in our study with 50.0% of case belonging to the pediatric age group. Most of the glaucoma in SWS presents unilaterally, though bilateral glaucoma has also been reported In our series,[16] 100% patients manifested unilateral glaucoma.

In cases of glaucoma, control of progression of glaucoma was difficult. Medical treatment, however, should be considered as the initial treatment of

choice Prostaglandin analogues should be used with caution because it can induce uveal effusion.[18-20]

Different surgical procedures have been performed in uncontrolled cases including cyclodestructive procedure, goniotomy, and trabeculectomy. Molteno or Ahmed implants may be necessary if these fail or even as the first intervention.[21]

Although the maximum dose of topical antiglaucoma drug was used for our 2 patients, trabeculectomy was needed in one eye for further intraocular pressure control at a later stage.

Patients with SWS suffer from variety of neurologic abnormalities, including epilepsy, hemiparesis, mental retardation, attention-deficit hyperactivity disorder, migraine, headache, and stroke-like episodes.

Epilepsy is the most important of the neurological alterations potentially associated with this syndrome. Seizures are frequently the first symptom of SWS. Twenty three to 89% of children with SWS have seizures.[22] While seizures may occur at any age, onset usually occurs in early childhood. In our study 83.33% patients presented with seizures as neurological manifestation.

The age of presentation of convulsion and their evolution is variable. In a retrospective case series, the onset of seizure ranged from birth to 23 years, and 12% of patients did not develop seizure until the third decade of life.[23] In our study also one patient did not present with convulsions till the age of 50 years.

Children whose seizures begin before the age of two years or intractable seizure with antiepileptic drug may eventually lead to motor deficits and mental retardation.[24] Mental retardation was seen in 33.33% of our patients.

Fortunately, Most of our patient who presented with seizure could be controlled with anti-epileptic drug.

Headaches affect 30 to 60% of patient with SWS.[25] In this present study, however, we found only 25% of patients had headache. This may result from the small number of cases collected in the present study or the rather young age of patients studied.

Leptomeningeal angioma may predispose them to neuronal hyperexcitability, causing changes in cortical perfusion and oxygenation consistent with theories on pathogenesis of migraine.[26]

The intracranial leptomeningeal angiomatosis is a key diagnostic feature in SWS.

The characteristic radiologic features are vascular abnormalities that commonly located in the parieto-occipital area.

Leptomeningeal angiomatosis manifests in 10% to 20% of the cases with typical facial angioma, generally on the same side. In our case series 25% patients were diagnosed to have leptomeningeal angiomatosis.

Both cerebral atrophy and cortical calcifications are considered to be an indirect consequence of chronic ischemia of the cortex due to vascular stasis in the area of leptomeningeal angioma.[27]

Plain skull X-rays illustrate the classic “tram-line” or “tram-track” calcification but are helpful only in diagnosis of SWS late in life.

Computed tomography provides adequate evaluation of brain calcification. Computed tomography showed cerebral calcifications in 50% of our patients.

MRI with contrast is the preferred and most sensitive imaging technique for evaluation of the leptomeningeal angiomatosis.[27]

CONCLUSION

Port-wine stains, glaucoma, and epilepsy were the most frequent clinical features of patients with SWS in the present study. When a patient presents with PWS covering the facial area of V1 distribution, MRI should be performed to screen for leptomeningeal angiomatosis and complete ocular examination should be carried out to screen for ocular complication especially glaucoma.

REFERENCES

1. Manivannan N, Gokulanathan S, Ahathya RS, Gubernath, DanielR, Shanmugasundaram. Sturge—Weber syndrome. J PharmBioallied Sci. 2012;4 Suppl. 2:S349—52.
2. Comi AM. Presentation, diagnosis, pathophysiology, and treat-ment of the neurological features of Sturge—Weber syndrome.Neurologist. 2011;17:179.
3. Pascual Castroviejo I. Síndrome de Sturge Weber. In: Pas-cual Castroviejo I, editor. Diagnóstico clínico-radiológico enneurología infantil. Barcelona: Ed. Científico-Médica; 1971. p.81—7.
4. Sullivan T.J., Clarke M.P., Morin J.D.: The ocular manifestations of the Sturge-Weber syndrome. J PediatrOphthalmol Strabismus 1992; 29:349-356.
5. Pascual-Castroviejo I., Diaz-Gonzalez C., Garcia-Melian R.M., et al: Sturge-Weber syndrome: study of 40 patients. PediatrNeurol 1993; 9:283-288
6. Sharan S, Swamy B, Taranath DA, Jamieson R, Yu T, et al. (2009) Portwine vascular malformations and glaucoma risk in Sturge WeberSyndrome. JAAPOS 13: 374-378.
7. Baselga E. Sturge-Weber syndrome. Semin CutanMed Surg 2004; 23: 87-98.
8. Luke RR, Malik SI, Hernandez AW, Donahue DJ, Perry MS. Atypi-cal imaging evolution of Sturge—Weber syndrome without facialnevus. Pediatr Neurol. 2013;48:143—5
9. Tallman B, Tan OT, Morelli JG, Piepenbrink J,Stafford TJ, Trainor S, et al. Location of port-wine stains and the likelihood of ophthalmic and/or central nervous system complications. Pediatrics1991; 87: 323-7.
10. Ch'ng S, Tan ST. Facial port-wine stains – clinical stratification and risks of neuro-ocular involvement.J Plast Reconstr Aesthet Surg 2008; 61: 889-93.
11. Baselga E. Sturge-Weber syndrome. Semin CutanMed Surg 2004; 23: 87-98.
12. Piram M, Lorette G, Sirinelli D, Herbreteau D,Giraudeau B, Maruani A. Sturge-Weber syndrome in patients with facial port-wine stain. PediatrDermatol 2012; 29: 32-7.
13. Sullivan TJ, Clarke MP, Morin JD. The ocular manifestations of the Sturge-Weber syndrome. J Pediatr Ophthalmol Strabismus 1992; 29: 349-56.

14. Di Rocco C, Tamburrini G. Sturge-Weber syndrome. Childs Nerv Syst 2006; 22: 909-21.
15. Sullivan TJ, Clarke MP, Morin JD (1992) The ocular manifestations of the Sturge-Weber syndrome. J Pediatr Ophthalmol Strabismus 29: 349-356.
16. Sharan S, Swamy B, Taranath DA, Jamieson R, Yu T, et al. (2009) Portwine vascular malformations and glaucoma risk in Sturge WeberSyndrome. JAPOS 13: 374-378.
17. Parsa CF (2008) Sturge-Weber syndrome: A unified pathophysiologic mechanism. Curr Treat Options Neurol 10: 47−54.
18. Awad AH, Mullaney PB, Al Mesfer S, Zwaan JT.Glaucoma in Sturge-Weber syndrome. J AAPOS 1999; 3: 40-5.
19. Yang CB, Freedman SF, Myers JS, Buckley EG,Herndon LW, Allingham RR. Use of latanoprost in the treatment of glaucoma associated with Sturge-Weber syndrome. Am J Ophthalmol 1998;126: 600-2.
20. Gambrelle J, Denis P, Kocaba V, Grange JD.Uveal effusion induced by topical travoprost in a patient with Sturge-Weber-Krabbe syndrome. J Fr ophtalmol 2008; 31: e19.
21. Hamush NG, Coleman AL, Wilson MR. Ahmed glaucoma valve implant for management of glaucoma in Sturge-Weber syndrome. Am J Ophthalmol 1999; 128: 758-60.
22. Pascual-Castroviejo I, Pascual-Pascual SI,Velazquez-Fragua R, Viano J. Sturge-Weber syndrome: study of 55 patients. Can J Neurol Sci2008; 35: 301-7.
23. Sujansky E, Conradi S. Outcome of Sturge-Weber syndrome in 52 adults. Am J Med Genet 1995; 57:35-45.
24. Roch ES, Bodensteiner J. Neurologic manifestations of Sturge-Weber syndrome. In: Bodensteiner JB,Roch ES, editors. Sturge-Weber syndrome. Mount Freedom, NJ: The Sturge-Weber Foundation;1999: 27-38.
25. Klapper J. Headache in Sturge-Weber syndrome.Headache 1994; 34: 521-2.
26. Lauritzen M. Pathophysiology of the migraine aura. The spreading depression theory. Brain 1994;117 (Pt 1): 199-210.
27. Baselga E. Sturge-Weber syndrome. Semin CutanMed Surg 2004; 23: 87-98.

How to cite this article: Shukla R, Baranwal P, Thakur AS, Rai G. Analysis of Sturge Weber Syndrome: A Retrospective Study. Ann. Int. Med. Den. Res. 2018; 4(5):OT10-OT13.

Source of Support: Nil, **Conflict of Interest:** None declared