

Introduction

Sturge – weber syndrome or encephalo trigeminal angiomatosis is the most frequent disease among the neurocutaneous syndromes and its proportion is 1/50,000 births. Clinically most specific and common finding of the SWS is the presence of naevus flameus also known as port wine stain. In addition to this other manifestations may be present, mainly those regarding changes to the central nervous system, such as convulsive crisis and mental retardation resulting from the leptomeningeal angioma several ocular manifestations can occur on ipsilateral side which include glaucoma (30%) choroidal haemangioma (40-50%), buphthalmos, prominent and tortuous conjunctival and episcleral vascular plexus affects 70% of cases, conjunctival and episcleral haemangiomas, heterochromia of the iris, angle abnormalities, optic atrophy etc. We report a case in which the patient do not have facial portwine stain but most other eye and CNS manifestation.

Case report

11 year old girl presented to us with complaints of swelling in the left eye. It was present since birth and it was not growing in size. Parents gave history of neonatal seizures involving right side of body and face. General examination showed alopecia aereta. All system examinations were normal with grade 5 power of both upper and lower limbs. Ocular examination of right eye showed heterochromia of the iris with otherwise normal anterior and posterior segment. Best corrected visual acuity of right eye was 6/6. Left eye showed nasal and temporal haemangiomas of size 0.5x1 cm. (Figure 1) Slit lamp examination detected dilated conjunctival and episcleral vessels along with heterochromia of the iris. Fundus examination showed diffuse choroidal haemangioma in the post pole extending from above the optic disc to the equator, veins found dilated. (Figure 2)Optic disc pale and cup was (CD0.3) found normal, tonometry detected intraocular pressure 20.6 mm of Hg in the left eye. Humphrey field analysis showed homonymous hemianopia. Best corrected visual acuity of left eye was 4/60. Gonioscopy and USG B scan of orbits were normal. USG B scan detected hyper echoic choroidal lesion. (Figure 3) Excision biopsy of the conjunctival mass confirmed haemangioma.

MRI brain with contrast showed leptomeningeal angiomatosis. CT scan showed characteritic ‘tram-line’ calcifications in the parieto – occipital cortex with unilateral cerebral atrophy, calvarial thickening, enlarged ventricles all suggestive of Sturge – Weber syndrome. (Figure 4 A,B,C,D)

Discussion

Meningo / Encephalo facial angiomatosis with cerebral calcifications - This condition is termed as Sturge – Weber syndrome since W Allen Sturge (1879) described a clinical case with focal seizures and facial portwine – stain

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Figure 4 A,B, C, D

and in 1922 Park Weber gave radio graphic evidence of atrophy and calcifications of cerebral hemisphere.

The clinical manifestations of Sturge – Weber syndrome have a common embryological basis, due to the developmental insult which affects tissues which originate in the pro and mesencephalic neural crest. So this is a disorder of neural crest migration and differentiation giving rise to vascular and tissue malformations in the meninges eye and brain.

Roach classified encephalofacial angiomatosis into 3 types based on varying degree of involvement.

Type I classic all the three manifestations namely facial, ocular and cerebral present.

Type II only facial manifestations and glaucoma are present in this type.

Type III This type manifest only with lepto meningeal angiomata.

Ocular manifestations

Choroidal haemangioma is seen in 40-50% cares of Sturge Weber syndrome and always unilateral and ipsilateral to the port wine stain.

- Extent and character of the choroidal vascular lesion result in striking reddish glow to the fundus known as "tomato catsup" fundus
- Hypermetropic refractive error can occur
- Retinal detachment can occur as a complication
- Prominent and tortuous episcleral and conjunctival vascular plexus affects 70% of cases and are after co-relates with increased episcleral venous pressure.
- Glaucoma is seen in 30-70% of patients and is almost always ipsilateral to the port wine stain, 20-30% cases are bilateral

- Other manifestations are conjunctival and episcleral haemangioma, heterochronia of the iris, angle abnormalities, optic atrophy, CRVO, ciliary body and orbital haemangiomas.
- Homonymous hemianopia also can occur (44%)

Systemic features are seizures (72-93%) hemiparesis (25-56%), headache (44-62%) developmental delay (50-75%)

Bilateral brain involvement is seen in 15%.

Management

Investigation

- X ray skull to show calcifications
- CT scan of the skull to show calcifications, vein abnormalities, brain atrophy
- MRI to show angiommas
- EEG to evaluate seizures

Treatment

Seizures: drug therapy may be used to control seizures, if resistant to treatment early surgical removal of the part of brain with the abnormal vessels may be considered.

- Photodynamic therapy for choroidal haemangioma
- Developmental delay – wide range of treatment options available
- Hemi paresis – Physical and occupational therapy

Prognosis

Prognosis depends on the specific neurological abnormalities present, eventhough some may worsen with age, SWS in not fatal disease.

Differential diagnosis


Conclusions

The case seen is our hospital is a rare case of sturge weber syndrome in which most of the ocular manifestations are present in one eye with absence of facial port wine stain, which requires a multidisciplinary approach by an Ophthalmologist, neurologist and a paediatrition, in order to achieve the best possible functional and cosmetic results.

References

3. Adams Textbook of Neurology