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Case report

Sturge-Weber syndrome: A case report

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Abstract

Sturge-Weber disease is characterized by a wine-colored nevus or hemangioma over the face in the ophthalmic division of the trigeminal nerve. Intracranially, hemangiomatosis often calcified occurs over the cerebral cortex on the same side as the facial lesion and usually in the posterior parietal occipital area. This report describes about 13 year old child with this disorder.

Key words: Emangioma, encephalotrigeminalangiomatosis, mother spot, SWS, Sturge-Weber syndrome

Introduction

Sturge-Weber syndrome (SWS) also called encephalotrigeminalangiomatosis, belongs to a group of disorders collectively known as "mother spot" and is found worldwide. Frederick Parker Weber, an English dermatologist first reported the radiologic features of brain atrophy in the disease named with William Allen Sturge in 1992. This congenital, non-familial disorder has become progressively less mysterious with the analysis of large series of patients accumulated over time (Oakes,1992).

Case report

A 13-year-old female first born child was admitted to the hospital with complaints of headache, weakness, and an episode of seizure followed by bed wetting, lack of orientation and loss of consciousness. The child was admitted in paediatric ward. After admission, the child had an attack of seizure. The MRI showed pial-angioma in the left parieto-temporal region, subcortical calcification in tram track pattern, left-sided hemiatrophy, a known case of SWS. With Hb level at 10.9gm% and ESR 49mm/hour, the child was obese with BMI of 25.2 kg/m^2 and bodyweight of 46kg. She was still in fourth standard and poor in studies because she was not able to write English alphabets in correct order. Doctors advised frequent eye checkups due to high risk of glaucoma. Now, the child has no special complaints and is stable.

Apparently, the child was normal up to four years of age. At the age of four years, the child was admitted with complaints of vomiting, drowsiness and an attack of seizure with bed wetting. MRI Brain showed features of SWS, child was treated and become normal and got discharged. There was no further treatment for the child because child was apparently normal after discharge. Again at nine years old, she had another episode of seizure (no jerky movements can be identified only by bedwetting), vomiting, fever and weakness of right extremities. She was admitted in PICU and treated with medications like T. Encorate, some antibiotics and physiotherapy. After this, the child had recurrent attacks of seizure. Seizure used to start with headaches. The child developed seizures at least once in two months. She is continuously on medications like T. Encorate 300mg OD, T. Carbamezapine 200 mg BD and was apparently normal even though there were recurrent attacks of silent seizures which lasted for 5-10 minutes, as reported by her mother.

During her antenatal period, the mother had attended regular antenatal checkups and the child was born by normal vaginal delivery. The child had Port Wine Stain (was pink in color and now became purple) on the left side of the face. During neonatal period, the

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baby was kept in NICU for observation. Breast feeding was started on the same day of delivery and had good sucking reflex. No abnormalities were reported during newborn period.

Description of disease

Sturge-Weber syndrome is a neurological disorder named after Dr William Allen Sturge (1850-1919) and Dr Federick Parkes Weber (1863-1962). It is defined by a facial port wine nevus and ipsilateralleptomeningealangiomatosis (Anjaiah, 2006). The port wine stain often involves V_1 and V_2 distribution of the trigeminal nerve. Majority of children with facial port wine stains do not have SWS, especially if they are limited to the midline of the forehead ('stroke-bite' birth mark). However, involvement of the upper and lower eyelid has a greater association with leptomeningealangiomas than involvement of the upper eyelid alone. Port wine stains may also involve the trunk and extremities. These lesions are present at birth and become hyper pigmented and larger with age. Lerptomeningealangiomas occur ipsilateral to the facial angiomas (Florine, 2011).

Incidence

It is one of the rare congenital disorders that occurs 1:20000-50000 live births. It is estimated that 1:1000 livebirths are born with port wine stain and only 6% of those babies are symptomatic (National Organization for Rare Disorders, 2003).

Etiopathogenesis

It is not a hereditary disorder, due to the random mutation in the GNAQ gene (G Protein Subunit Alpha Q) is a protein coding gene (Comi, 2003).

It is estimated that this occur due to the formation of abnormal blood vessels during fetal life. As a result, the circulation to brain leads to re-organization, but the ectoderm differentiates into skin of the upper face and occipital lobe. The superficial leptomeninges become highly vascularized and the part of the brain under this area becomes atropic and calcified.

Types

Sturge-Weber syndrome is mainly of three types:

- *Type I:* Includes facial and leptomeningealangiomas, risk for glaucoma or choroidal lesions, unilateral, and most common type.
- *Type II:* Includes facial angioma, risk for glaucoma, no brain involvement. Symptomatic at any time beyond port wine stain. Glaucoma, headache, and cerebral blood flow abnormalities are symptoms.
- *Type III:* Includes exclusive involvement of leptomeningealangioma, absence of facial angioma, rarely glaucoma. It can be diagnosed only through brain scan. Port wine stain on forehead, upper eyelid may be bilateral or unilateral. (Anjaih, 2006)

In the present case child has type II SWS, because child has headache, port wine stain, and risk for glaucoma.

Clinical features

- Port wine stain: - The most common, red-pink in colour mostly unilateral. (Wong,1997)
- Developmental delays
- Seizures: - 75-90%
- Cognitive impairment: - 50-70%
- Paralysis (hemiparesis): - 25-60%
- Hemianopsia: - 40-45%
- Weakness
- Muscle weakness
- Vascular headache: - 40-60%
- Buphthalmose
- Glaucoma
- Vision impairment
- Sensitivity to light
- Eye pain
- Learning disabilities
- Intracranial calcification

It can be also classified as following: (Anjaiah, 2006)

I. Cutaneous features

- Classically, the cutaneous angioma in SWS is found unilaterally on the forehead or about the eyes. Sometimes the whole face and body may be involved. It is due to the persistence of embryonic venous plexus. There is a traditional link between the nevus and trigeminal nerve.

II. Ophthalmic complications

I. Glaucoma is present in 42% of cases. Bilateral glaucoma is possible. Homonymous hemianopia is present.

III. Neurologic manifestations

- Most patients will have normal function
- Epileptic seizures and hemiparesis may be present

- Seizures are present in 72% of cases with unilateral involvement and in 93% patients with bilateral involvement.
- If seizures start below two years of age, it suggests greater chance of mental retardation and refractory epilepsy.
- Hemiparesis
- Arrested growth of extremities
- Communicating hydrocephalus is a rare complication

All children with SWS have port wine stain, but all with port wine stain has no SWS.

In the present case child has seizures, vascular headache, at risk for glaucoma and has the history of hemiparesis.

Diagnostic measures
- History collection
- Detailed physical examination with neurological assessment (port wine stain as a birth mark)
- Laboratory studies of blood
- MRI (Magnetic Resonance Imaging)
- CT (Computed Tomography)- tram track calcification
- Ophthalmic tests

In the present case, the child had pink port wine stain from the birth onwards and was pink in colour. Ophthalmic checkups were done frequently and C T Brain shows SW Syndrome.

Management

Most of the time the treatment is symptomatic:
- Anticonvulsants
- Surgeries like goniotomy, trabeculotomy for the treatment of glaucoma
- Eyedrops like prostaglandins, beta-blockers, carbonic anhydrase inhibitors for reducing the intraocular pressure.
- Analgesics in case of vascular headache
- Educational therapy
- Physiotherapy for strengthening of muscles
- Pulsed dye laser therapy achieves significant lightening and may prevent the hypertrophy or vascular nodules, which can develop later on in life. Laser therapy, with a wavelength of 585-600 mm, targets oxyhaemoglobin, leading to destruction of ecstatic vessels without significant damage to surrounding skin structures (Kyle, 2009).

  In the present case, the child was on medications like T. Encorate300mg OD, T. Carbamezapine 200 mg BD and was instructed for regular ophthalmologic examination.

Complications
- Glaucoma
- Partial epilepsy
- Headache
- Stroke like episodes
- Neuro-behaviour

In the present case, the child had the history of hemiparesis, headache, epilepsy, and the risk for glaucoma.

Foundations and support groups

To improve the quality of life among children with SW syndrome foundations and support groups are available internationally. They are celebrating SW Awareness day on November 1st; birthday of Dr William Allen Sturge.

Discussion

In the above-mentioned case, even though the child has port wine stain from birth onwards, it remained asymptomatic up to four years. Early detection of the disease helps the child to manage the disease effectively and remain free from dangerous complications. However, it is essential to monitor the long-term side effects of anticonvulsants because child is obese. Moreover, it is important to provide academic support and proper attention to the child by provision of activities which increase concentration and help her studies. In fact, in this case eye checkups are mandatory in order to detect the visual problems at an initial stage. The child is treated effectively with the help of medicines and the seizures lessen over time. Due to early diagnosis and treatment, the child has good prognosis.

Conclusion

Sturge-Weber syndrome is a neurocutaneous disorder of facial port wine stains and vascular malformations of the leptomeninges and eye. It includes the following triad-* port wine stain involving at least the V1 branch of the trigeminal nerve,* ipsilateralleptomeningealangiomatosis and *ipsilateral vascular malformation of the choroidalvaculature of

the eye (Florine, 2011).The child has the history of hemiparesis. She has port wine stain which was pink at birth and now became purple. She has the risk for glaucoma, has recurrent attacks of seizures, headache and is under anti-convulsant therapy.

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