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Sturge Weber Syndrome - Roach's Type II Variant

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Abstract

Sturge-Weber syndrome (SWS) is a neurocutaneous sporadic disorder caused by mutation in GNAQ gene responsible for persistence of vascular plexus around cephalic portion of neural tube. It has a wide spectrum of cutaneous, neurologic and ophthalmic manifestations, which may or may not be associated with one another. Roach scale has classified it into three types. Here, we present a case of Roach's Type II variant of SWS with Port-wine stain (PWS) and ocular abnormalities without Central Nervous System (CNS) involvement. A 24 months old female presented with hemangioma involving the left side of face since birth. She had history of corneal edema and buphthalmos at two days of life. There was no history of seizure or developmental delay and Magnetic Resonance Imaging (MRI) of the head ruled out cranial hemangioma. Roach's Type II is a rare variant of SWS and should be suspected in any case having PWS along the course of trigeminal nerve with congenital glaucoma because the neurologic involvement in a given case may vary from an absence to overt clinical manifestations with or without radiological changes. Due to its wide range of manifestations, a multidisciplinary approach is required for proper management of these patients.

Key words: Neural Tube; Neurocutaneous Syndromes; Port-Wine Stain; Sturge-Weber Syndrome

Introduction

Sturge-Weber syndrome (SWS) or encephalotrigeminal angiomatosis is a neurocutaneous disorder caused by somatic mutation in GNAQ gene leading to residual embryonal blood vessels.¹ During the sixth week of intrauterine life, a vascular plexus develops around the cephalic portion of the neural tube, which normally regresses at around ninth week of gestation. In SWS there is failure of this normal regression of vascular plexus leading to angiomatous formation.² It has an incidence of 1 per 50,000 live births and most of the cases are sporadic with no gender predilection.^{3,4}

The diagnosis of SWS is made based on cutaneous, ocular and CNS abnormalities. The cutaneous abnormality manifests as facial angioma involving the trigeminal nerve distribution.⁵ Ocular abnormalities

include glaucoma, choroidal hemangiomas and heterochromia of irides. Neurological manifestations include seizures, hemiparesis, intellectual disability and learning difficulties caused by leptomeningeal angiomas (LA).⁶

The Roach Scale is often used for the classification of SWS.⁷

Type I - Facial and LA; may have glaucoma

Type II - Facial angioma alone; may have glaucoma

Type III - Isolated LA; usually no glaucoma

Here, we present a case of Roach's Type II variant of SWS with PWS and ocular abnormalities without CNS involvement, a rare presentation.

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Case Report

A 24 months old female child presented with hemangioma involving the left side of the face since birth. Further history revealed that she had corneal edema and buphthalmos diagnosed at two days of life with improvement at one month of age, after treatment from ophthalmological side. At the age of three months she started developing facial asymmetry with increase in the diameter of left side of the face. At the same age, ocular examination revealed choroidal hemangioma with normal optic disc. At five months of age, MRI of the head was performed which excluded any cranial hemangioma. However, she started developing bleeding from the upper front region of the mouth from the age of 13 months which confirmed vascular malformation over the left side of the maxilla being impinged by the erupting tooth on dental examination. On examination, port-wine stain was seen over the left side of V1 and V2 region face with extension to the ipsilateral scalp. However, there was no history of seizure or developmental delay in the child. The parents were advised for regular follow-ups of the child and the treatment options were explained. The child has been planned for further intervention in future at appropriate age taking into consideration the parents' opinion.

Discussion

SWS is a rare neurocutaneous disorder belonging to the Phakomatoses group of disorders with constellation of features of PWS, LA, ocular changes and sometimes extracranial angiomas and soft tissue overgrowth.⁸ Presence of both CNS and facial angiomas denotes complete SWS whereas absence of any of the two forms incomplete SWS and the Roach classification can be used for this⁸

The characteristic port-wine stain also known as nevus flammeus is present over the face along the trigeminal

nerve distribution, of which ophthalmic division is the most commonly involved. The color of this PWS vary from pink, red to purple and size from small to large macular lesions which usually do not cross the midline.⁹ Approximately 85% has unilateral and 15% has bilateral involvement, and 68% had involvement of more than one dermatome. CNS or eye involvement is commonly seen in those individuals with V1 and V2 involvement.¹⁰ However in about 13% of individuals with diagnosis of SWS, facial angioma cannot be detected which falls in Type II variant of Roach Scale.¹¹

Neurologic dysfunction results from hypoxia, ischemia, venous occlusion, thrombosis, infarction, or vasomotor phenomenon developing around the angioma which can later progress to calcification, gliosis, and atrophy. All these lead to clinical manifestations of seizures, headache, hemiparesis and developmental delay.¹² The incidence of seizures in patients with SWS is 72-93%, headache is 44-62%, hemiparesis is 25-56% and developmental delay and mental retardation is 50-75%.¹¹

The ocular manifestations are produced secondary to mechanical obstruction of the angle of the eye, elevated episcleral venous pressure, or increased secretion of aqueous fluid. The incidence of glaucoma in SWS ranges from 30 to 71%. It may be present at birth but can develop at any age in adulthood. Similarly, the incidence of choroidal haemangioma in SWS is 40%.

Vascular selective lasers are employed for the treatment of PWS of which Pulsed Dye Laser (PDL) is the most commonly used treatment modality. It uses the principle of selective photo-thermolysis in which the affected cutaneous blood vessels are targeted protecting rest of the skin. Multiple sessions of therapy may be required but there is no uniformity in the ideal age for initiation of the treatment. The chances for better outcome increase with decrease in age of



Fig 1a: At birth



Fig 1b: At 15 months



Fig 1c: At two years

the patient as children have thinner skin and more superficial vessels.

In a study conducted in 1998, the percentage decrease in the size of PWS depending on the age suggests that intervention is required early in life. In those <1 year of age, there was 63% improvement in after five treatments as compared to 1-6 years and >6 years age group having improvement of 48% and 54% respectively.¹³ Similarly, another study showed 100% clearance of PWS was higher in younger children as compared to the older. There was 100% clearance in 32% of children <1 year, 23% in 1-2 years, 17% in 2-6 years, 16% in 6-12 years and 18% in >12 years age group.¹⁴ However, there are limitations to use of PDL especially in infants and children. The most serious complication associated with PDL is ocular damage resulting from laser radiation. PDL is association pain and topical anesthesia when applied to large surface area or general anesthesia for young children may have risk. Also, immobilization of young children may become an issue. So, delaying elective procedures until a patient is at least 3 years of age can be considered.¹⁵

In our case the patient complained of the port wine stain and history of ocular involvement in the form of both glaucoma and choroidal hemangioma without any history suggestive of CNS involvement. No abnormalities could be appreciated in the MRI of brain. SWS may have incomplete presentation as seen in our case which falls under Type II variant of Roach Scale. This type of presentation of SWS is very rare. Hence, the diagnosis of SWS should not be excluded in absence of classical triad of central nervous system, ocular and cutaneous involvement.

Conclusion

SWS is an angiomatosis commonly presenting with the triad of PWS, ocular abnormalities and LA. However, diagnosis of SWS cannot be excluded in absence of any one of the components according to the Roach classification. Here we present a case of SWS in absence of leptomeningeal angiomas which is a Roach Type II variant of SWS. Also, it is important to emphasize the requirement of a multidisciplinary team for proper management of these patients.

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