

Case Report

Sturge-Weber Syndrome in A Neonate: An Unique Presentation

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Abstract: The Sturge-Weber Syndrome, also known as encephalotrigeminal angiomatosis, is a rare vascular neurocutaneous syndrome. It is characterised by port wine stain, seizure, and intracranial calcification. The facial nevus follows the trigeminal nerve in the face. We report a case of Sturge-Weber Syndrome presented in early neonatal period presenting with microcephaly, cranial synostosis, seizure and bilateral port wine stain.**Keywords:** Sturge-Weber Syndrome, portwine stain, tram-track calcification

INTRODUCTION

Sturge-Weber syndrome is a rare developmental capillary malformation originating during embryogenesis. Defect lies in neural crest migration. Anomalous development of the primordial vascular bed in the early stages of cerebral vascularization is the cause of this disease. It was first described by Schirmer in 1860 and later more specifically by Sturge in 1879. The classic feature of this disorder is the angioma of leptomeninges. Most common features are Port-wine stain and dermal angiomas, intracranial calcification, mental retardation, seizure, ocular involvement. The facial angiomas are generally seen in maxillary and ophthalmic area of trigeminal nerve distribution. The first sign of presentation was vascular features. Developmental delay is more marked if facial involvement is bilateral. Cranial CT shows characteristic Tram-Track appearance. Generally the diagnosis is delayed due to late appearance of radiological features. Hence very rarely reported in neonatal period.

We present a case of Sturge-Weber syndrome in an early neonate with characteristic clinical and radiological pictures.

CASE REPORT

A near term neonate with gestational age 36 weeks was presented to the neonatal emergency with chief complaint of abnormal movements and abnormal cry after birth. APGAR score was 7,9 respectively in 1 and 5 minutes. Birth weight was 3550 grams. Head circumference was 30 cms. Length was 52 cms and abdominal circumference was 32 cms. Upper segment and lower segment were 28 and 23 cms. On examination baby was pink alert, with feeble cry, decreased activity, closed fontanelles, high arched

palate, facial redness, patent anus. Baby had normal female genitalia. Systemic examinations were within normal limits. Baby was admitted, given IV fluid according to institutional protocol. She was treated by IV ampicillin and amikacin. On 30th hour of admission baby again developed seizure, which was subtle in nature and associated with cycling movements. Baby was given IV phenobarbitone loading followed by maintenance. Seizure subsided. She improved with normal cry and good sucking reflex. She was given direct breast milk on fourth day. It was observed on day 4 while crying upper portion of face (upper portion of eye brow both sides) was looking red (fig-2). Upon observation it was found to be bilateral portwine stain. A NCCT scan was done. It showed multiple gyrus calcification bilaterally with mild hydrocephalus and basal ganglia calcification (fig-1,3). The calcification had tram-track appearance. A torch screening was done to rule out the infection, it came as negative.

DISCUSSION

Sturge-Weber Syndrome is a congenital phakomatosis. It is presented with vascular malformations resulting from the failure of fetal veins to develop normally leading to changes in the brain, skin, and eye. These malformations cause venous hypertension and hypoperfusion of the underlying cortex causing cerebral ischemia, atrophy, and neurological deterioration. This syndrome occurs with equal frequency in both sexes, with seizures typically developing in the first year of life [1]. The inheritance of Sturge-Weber syndrome is sporadic and it occurs with a frequency of 1: 50,000 [2]. Sturge-Weber syndrome is a rare neurocutaneous disorder and is referred to as complete when both central nervous system and facial angiomas are present and incomplete when only one

area is affected without the other[3]. The Roach Scale is used for classification, as follows [4]:

Type I - Both facial and leptomeningeal angiomata; may have glaucoma

Type II - Facial angioma alone (no CNS involvement); may have glaucoma

Type III - Isolated leptomeningeal angioma; usually no glaucoma.

Neurological outcome in children with SWS is highly variable, ranging from minimal or no neurological signs to a devastating impairment with uncontrolled seizures, hemiparesis, visual field defect and progressive mental retardation. Seizures is a very common feature, often occurs during the first year of life [5]. The hypoperfusion of cortical tissue is further accelerated by seizures, thereby worsening the prognosis [6]. Neuroimaging studies help to establish the diagnosis, assess severity and follow the progression of brain involvement in SWS. Cortical atrophy

underlying the angioma with gyriform "tram track" calcification is the characteristic imaging feature. Calcification however is unusual before 2 years of age and most commonly involves the parietal and occipital lobes, but it is seen very rarely in newborns. The best imaging modality is MRI while calcifications can be assessed in detail on CT images[7]. Port wine stains in childhood are classically faint, pink macules, tend to darken progressively to red purple; may be isolated with well-delineated border or may be very diffuse (like our case). It has usually unilateral distribution along one or more segments of trigeminal nerve. Occasionally bilateral involvement or additional port wine lesions are found elsewhere in the body[8]. In our case, bilateral involvement of the face was seen. Vascular malformation of eye is seen when ophthalmic branch of trigeminal nerve is involved. Intraoral involvement is common, resulting in hypervascular changes to the ipsilateral mucosa [9].

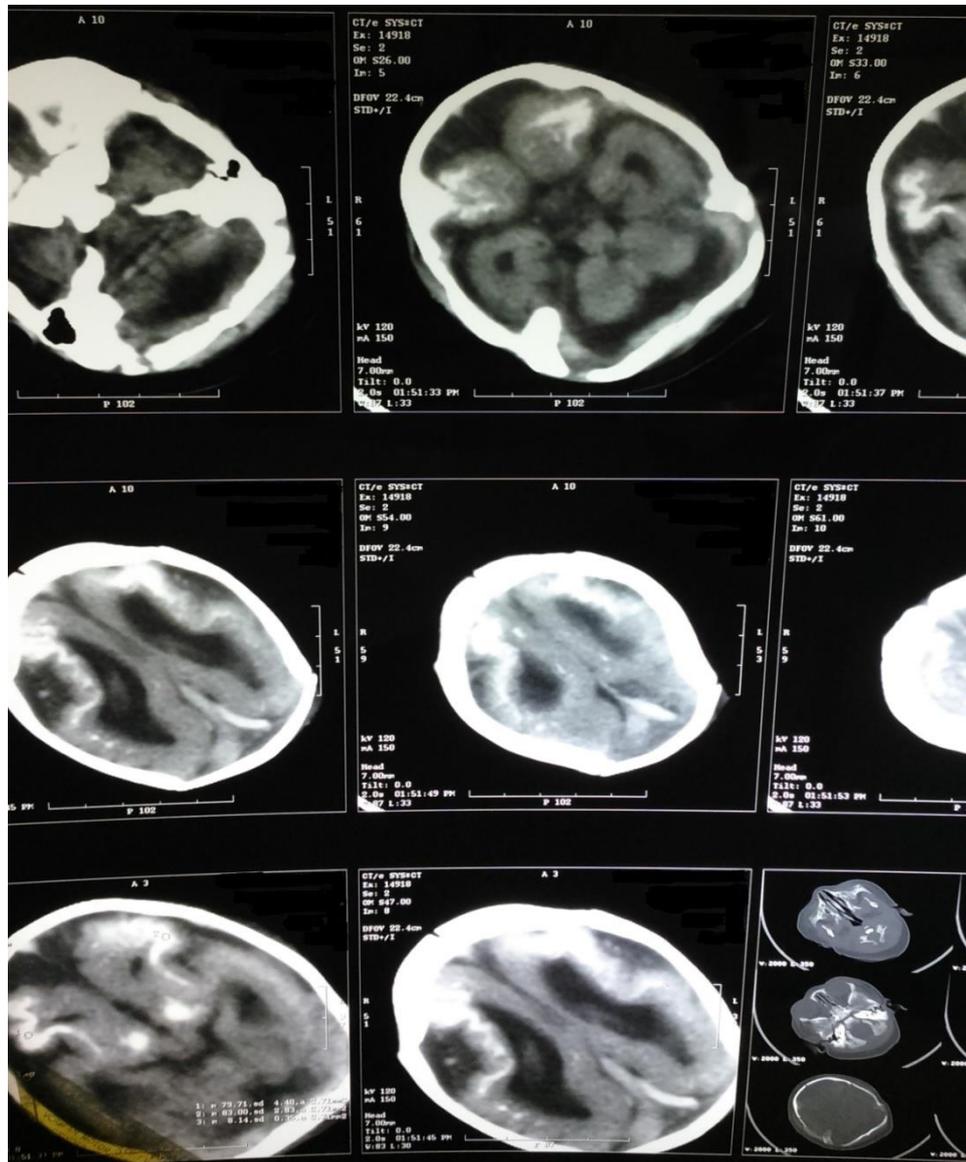


Fig-1: Bilateral tram track appearance



Fig-2: Bilateral PWS shown in arrow

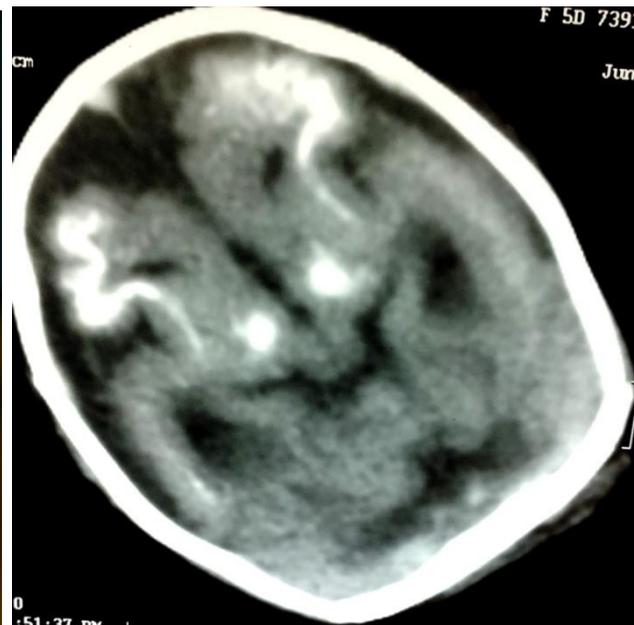


Fig-3: bilateral tram track appearance

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