

STURGE-WEBER SYNDROME: A CASE REPORTJANANI R^{1*}, KIRUTHIKA S², ARUNACHALAM RAMACHANDRAN³, KUMARESAN A²

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ABSTRACT

To report a clinical case of Sturge-Weber syndrome (SWS) 1 year 8-month-old male child reported to Saveetha Medical College, Physiotherapy Department presented with complaints of difficulty in sitting, standing, and walking. Computed tomography brain revealed unilateral calcification and cerebral atrophy on the right side. Physical diagnosis revealed the child had left hemiparesis with developmental delay. The child had a developmental delay with a motor developmental age of 2 months. As clinical manifestations of SWS are wide, it leads to multidisciplinary approaches. Physiotherapist's knowledge is important for early diagnosis and to provide an adequate physiotherapy treatment.

Keywords: Sturge-Weber syndrome, Hemiparesis, Developmental delay.

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INTRODUCTION

Sturge-Weber syndrome (SWS), or encephalotrigeminal angiomas, is a rare, congenital neurocutaneous syndrome that affects the meninges of the brain and the skin of the face. First described by Schirmer and later more specifically by Sturge in 1879, it is also known as Sturge-Weber disease, leptomeningofacial angiomas and Sturge-Weber-Dimitri syndrome 1. Involvement is usually unilateral. Embryonic blood vessels that fail to regress at the appropriate time of development is known to be the cause of this disease. As a consequence, residual blood vessels form angiomas on the face, in the meninges, and in the ipsilateral eye occurs [1]. The incidence of SWS is estimated to be 1/50,000 live births and no racial bias. The common manifestations related to central nervous system (CNS) are the CNS paresis, hemiparesis, developmental delay, and headache. Manifestations related to face are cutaneous angiomas called port wine stains that occur along dermatomes supplied by the ophthalmic and maxillary division of trigeminy the trigeminal nerve most often affected in the distribution of the trigeminal nerve. SWS belong to the group of the disorder known as phakomatoses ("mother-spot" diseases). Sturge associated dermatological and ophthalmic changes of the disease to neurological symptoms [2].

CASE REPORT

A 1 year 8-month-old male child reported to Saveetha Medical College, Physiotherapy Department with complaints of difficulty in sitting, standing, and walking.

The past medical history revealed that the child had an uneventful prenatal period. The child was born full term through normal vaginal delivery and cried immediately. His first episode of focal seizures was at 1st month of age with duration lasting for 2 min. He had 3 episodes of seizures. He was on antiepileptic drugs. The developmental history is as follows: The child attained partial head control at 8 months; whole grasp was attained at 5 months. Social smile not attained and does cooing sounds. computed tomography brain revealed unilateral calcification and cerebral atrophy on right side taken on 25th June 2017. In view of clinical neurological findings the diagnosis of SWS was then established.

On observation, the child had a unilateral port wine stain centered around right forehead, the right eye, side of the nose, right upper lip, palmar area of the right hand, and plantar surface of the right foot.

On examination of higher mental function child is conscious and irritable. The child does not respond to visual stimuli. Responds to an auditory stimulus such as mother's voice and rattles.

On motor examination, the child had reduced tone on the left upper limb and lower limb. Deep tendon reflexes were diminished, and a flexor response of Babinski sign was present on left side. All primitive reflexes were persistent. Moro and startle reflex were reduced on left side attributed due to weakness on left side. An assessment of Voluntary control, the child, did not cross the midline to reach objects using the left upper limb with no initiation of movement and kicking activity were reduced on a left lower limb. The child only initiated movements of the right upper limb and kicking was present in the right lower limb.

On a sensory examination, superficial sensation (assessed on body segments) was intact on the bilateral side of the upper limb and lower limb.

On developmental assessment, the child had mild head lag on pull to sit the test, lifts head up for a few seconds in prone position. In supine position, the child does not turn head to the left side. The child does not initiate rolling on both sides.

Physical diagnosis revealed the child had left hemiparesis with developmental delay. The child had a developmental delay with a motor developmental age of 2 months.

DISCUSSION

This study reports a child of SWS with hemiparesis and developmental delay. SWS is referred to as complete when both CNS and facial angiomas are present, and incomplete when only face or CNS is affected. SWS is classified as follows, according to Roach scale [2] Type I - facial and leptomeningeal angiomas; the patient may have glaucoma, Type II - facial angioma alone (no CNS involvement); the patient may have glaucoma, and Type III - isolated leptomeningeal angiomas; usually no glaucoma. According to Roach scale, this case is a Type I SWS. The vascular dynamics causing precipitation of calcium deposits in the cerebral cortex are altered by the presence of angina [3]. Vascular plexus starts developing during the 6th week of intrauterine life and usually undergoes regression during the 9th week. The persistence of vascular plexus around the cephalic portion of the neural tube is the

cause of SWS [4]. According to INAN (1999), 87–90% of the cases have port wine stains localized over the face on the right side. 50% of the patients have lesion extended over the median, and 33% of patients have bilateral involvement. In this case, the child also showed port wine stains only on the right side of the side without extension over midline [5].

The differential diagnosis includes rendu–Osler–Weber syndrome (Hereditary hemorrhagic telangiectasias), Maffucci's syndrome, angina osteodystrophy syndrome, von Hippel-Lindau disease, and Klippel-trenaunay-Weber syndrome. Isolated lesion such as nevus flammeus neonatorum should be differentiated from port-wine stains [6-10].

Most often a Flashlamp-Pulsed Tunable Dye Laser is used to reduce port-wine stains in children and treats the abnormal vascular structure beneath the skin [11]. These children may require surgery to remove the areas of brain tissue responsible for epilepsy. Surgical options for these children may include a complete hemispherectomy or a focal resection-based on the location of epileptic tissue [12]. Physical therapy intervention for SWS is focused on impairments associated with hemiparesis and developmental delay. Common physical therapy interventions given are muscle-strengthening, Orthotics education, spasticity management, and constraint-induced movement therapy [13]. For this child developmental training can be given for the delay and facilitator technique can be given to improve muscle tone on the hemiparetic side.

CONCLUSION

The treatment of the SWS depends on the presentation or clinical features. As clinical manifestations of SWS are wide, it leads to multidisciplinary approaches for its management, such as a pediatrician, Neuro physician, Ophthalmologist, Cosmetologist, Physiotherapist, radiologist, and dentist. As SWS is a rare congenital, developmental,

disorder manifesting with a facial port-wine birthmark, affecting the CNS, therefore, physiotherapist's knowledge is important for early diagnosis and to provide an adequate physiotherapy treatment.

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