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Case Report

Sturge Weber Syndrome on Computed Tomography: A Rare Case Report and Review of Imaging

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Abstract: Sturge-Weber syndrome (SWS), also called encephalotrigeminal angiomatosis, is a neurocutaneous disorder with angiomas that involve the leptomeninges and the skin of the face. It is a rare congenital non-hereditary disorder with incidence of 1: 20,000 – 50,000. Both sexes are affected equally. Presentation is usually in childhood. Imaging is the most useful investigation in evaluation of the disease. A 28-year woman was referred for brain CT. She presented to the maxillo-facial unit with complain of right facial swelling and discoloration since birth. There was history of developmental delay, recurrent headache and weakness of the left upper and lower limbs. There were occasional episodes of seizures. No history of loss of vision. On examination, significant findings were port-wine stain, swelling on the right side of the face and reduce power on the left upper and lower limbs. Cranial CT images revealed loss of right cerebral volume with extensive tram track calcifications involving predominantly the right parietal and occipital lobes a diagnosis of SWS was made. Patient is currently on medical management.

Keywords: Sturge-Weber Syndrome, Adult, Port-Wine Stain, Computed Tomography.

INTRODUCTION

Sturge-Weber syndrome (SWS), also called encephalotrigeminal angiomatosis, is a neurocutaneous disorder with angiomas that involve the leptomeninges and the skin of the face. It belongs to a group of disorders collectively known as the phakomatoses. It is a rare congenital non-hereditary disorder with incidence of 1: 20,000 – 50,000. Both sexes are affected equally and it shows no racial bias in distribution [1, 2]. The disorder usually presents in childhood with seizures, stroke like episodes, developmental delay and port wine stain of the face in the region of the ophthalmic and maxillary distribution of the trigeminal nerve [2].

Imaging is the most useful investigation in evaluation of the disease with computed tomography and magnetic resonance imaging playing a pivotal role in demonstrating the intracranial changes [3]. We present a 28 year woman diagnosed with sturge weber syndrome on CT scan with review of imaging.

CASE REPORT

A 28-year woman was referred to radiology department, Usmanu Danfodiyo University Teaching Hospital, Sokoto for brain CT. She presented to the maxillo-facial unit with complain of right facial swelling and discoloration since birth. There was history of developmental delay, recurrent headache and weakness of the left upper and lower limbs. There were occasional episodes of seizures. No history of loss of vision. She is not a known hypertensive, diabetic mellitus or sickle cell disease patient. On examination, significant findings were port-wine stain, swelling on the right side of the face and reduce power on the left upper and lower limbs. Cranial CT images revealed loss of right cerebral volume with prominence of the right cerebral sulci (fig. 1). There were extensive tram track calcifications involving predominantly the right parietal and occipital lobes with slight extension to the temporal and frontal lobes (fig. 1 and 2). The paranasal sinuses and orbits were normal in size. However, soft tissue swelling was noted on the right side of the face. A diagnosis of SWS was made. Patient is currently on medical management.

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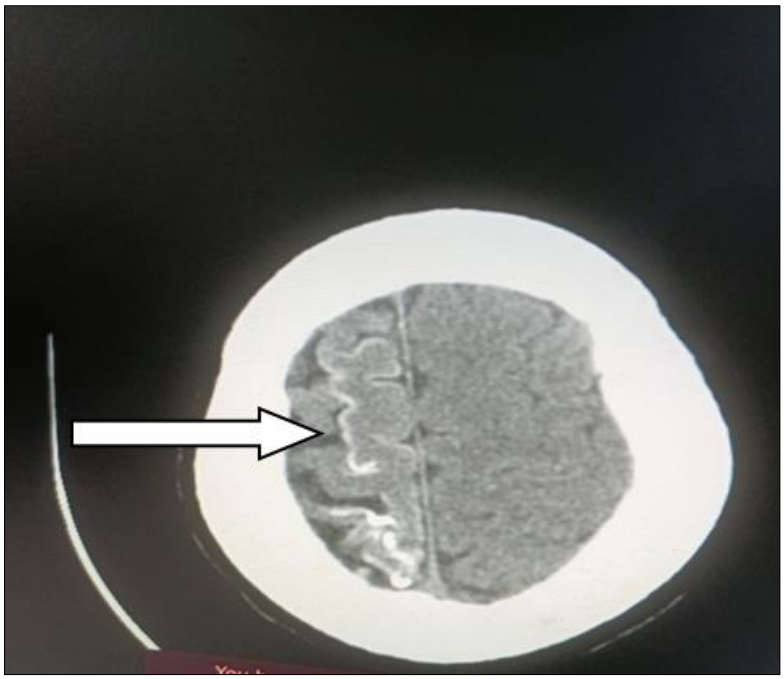

Figure 1: Non contrast enhanced cranial computed tomogram showing reduction in volume of right cerebral hemisphere (arrow) with areas of tram track gyral calcifications (HU = 126)

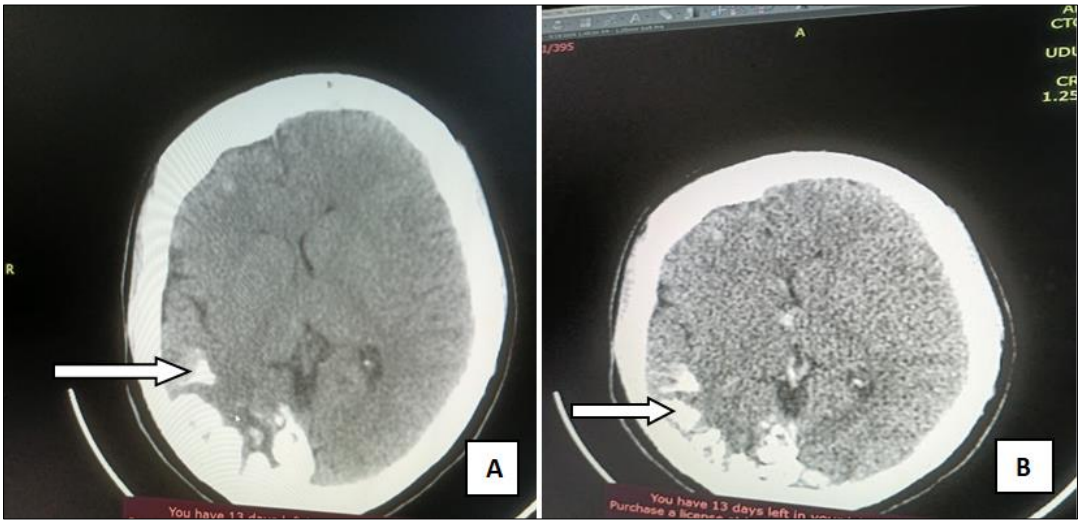

Figure 2: Non contrast computed tomogram (A) and contrast enhanced computed tomogram (B) of the brain showing areas of non enhanced gyral hyperdensity (HU = 126) in the right cerebral hemisphere (arrows)

DISCUSSION

Sturge–Weber syndrome (SWS) is a rare vascular disorder, first reported by William Allen Sturge in 1879 [4, 5]. It is characterized by the association of a facial birthmark called a port-wine stain involving the region of distribution of trigeminal nerve, neurological manifestations such as seizures, hemiparesis, intellectual disability, and learning difficulties; and ocular abnormalities which includes glaucoma, choroidal haemangiomas, or heterochromia of irides [6]. In addition to these typical manifestations, patients with SWS may present with endocrine abnormalities such as growth hormone deficiency and central hypothyroidism [7, 8]. The index patient presents with the history of seizure, hemiparesis and facial port-wine stain.

The diagnosis of SWS is based on characteristic clinical symptoms, facial appearance, and imaging findings [9]. Imaging modalities employed in the evaluation of patients with this disorder include magnetic resonance imaging (MRI), computed tomography (CT), positron emission tomography (PET) scan, Ultrasonography and plain radiography. The most common locations of brain abnormalities in SWS are the occipital, posterior parietal and temporal lobes[9]. This is also demonstrated in the index patient. Brain MRI (T1 and T2) with and without gadolinium contrast, susceptibility (SWI) and diffusion (DWI) weighted sequences are recommended [10]. The MRI findings will depend on the stage of the disease. In the early stage, imaging may be false-negative due to incomplete brain

myelination. However, in the late stage, an increased T2 signal is seen in the region of gliosis with decreased pial enhancement and cortical atrophy. Gyriform calcifications are best seen on T2 or SWI and appear as areas of signal void along the gyri in a serpentine pattern. A screening MRI should be considered in extensive birthmarks if presymptomatic treatment is considered [2-10].

Computed tomography is the best for detecting gyriform calcifications in the brain and also shows other changes, such as cortical atrophy and leptomeningeal enhancement after contrast administration. Some of these changes were demonstrated in the index case. The use computed tomography in children is not recommended due to the risk associated with ionizing radiation in ongoing brain development [9-11]. The PET scan though not routinely employed, may be a useful modality for studying cerebral metabolism in patients with SWS [2-9].

Skull radiographs may demonstrate gyriform (tram-track) calcifications, while ocular ultrasonography is useful in demonstrating ocular abnormalities such as choroidal hemangioma [9].

The differential diagnosis of SWS are blue rubber bleb nevus syndrome, meningoangiomatosis and Klippel-Trenaunay-Weber syndrome. Also include posterior fossa abnormalities, hemangiomas, arterial anomalies, cardiac, eye, and sternal anomalies (PHACES) syndrome and the Wyburg-Mason syndrome [9-12]. All these can be differentiated from SWS by the clinical and imaging features of SWS earlier described.

CONCLUSION

Sturge-Weber syndrome is a rare neurocutaneous disorder which commonly presents in childhood. This case is an unusual clinical presentation in adulthood and highlights the role of imaging in the evaluation of this disorder.

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