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Sturge–Weber Syndrome – A Case Report

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Abstract **Case Report**

Sturge-Weber syndrome (SWS) or encephalofacial angiomatosis is a rare neurocutaneous and congenital ocular syndrome. It is responsible about two malformations: congenital facial capillary planar angioma and leptomeningal venous-capillary angioma (most often parieto-occipital homolateral angioma). Neuroimaging, especially magnetic resonance imaging (MRI), makes the diagnosis, ideally before ocular complications. In Sturge-Weber syndrome neurodevelopmental outcomes depend to accurate recognition of the signs of severity and proper therapeutic management of epilepsy. We report the case of an old man in whom SWS was suspected based on facial angioma and epilepsy.

Keywords: Sturge-Weber, adult, planar angioma, CT scan.

INTRODUCTION

Sturge-Weber syndrome, also known as encephalotrigeminal angiomatosis, is a neurocutaneous affection characterized by angiomas involving the face, choroid, and leptomeninges. It is the most frequent phacomatosis after neurofibromatosis and tuberous sclerosis [3]. The neurologic manifestations of SWS include mainly seizures [3]. MRI is the primary imaging modality recommended for clinical practice. Other imaging modalities (CT, PET) have a limited role and can be used in other specific clinical or research investigations [1].

CASE REPORT

We report the case of 20-years-old men who consulted to the radiology department for seizures, delay mental development with notion of cranial trauma one day ago. No epilepsy was noted in the family history, no notion of consanguinity. On examination, the patient was conscious, apyretic, with no sensory-motor deficit. On clinical examination, a cutaneous planar angioma was found along the mandibular branch of the trigeminal nerve (cheeks); (Figure 1).

A cerebral CT scan was performed (Figure 2) showing thickening of the left choroid plexus, gyriform calcifications, and an accentuated gyriform contrast of the cortical sulci which are enlarged testifying to this homolateral cerebral atrophy. Therefore, in view of all the clinical and radiological signs, the diagnosis of Sturge Weber syndrome was retained.

DISCUSSION

Sturge-Weber syndrome (SWS) is a neurocutaneous disorder associated with facial port wine birthmark, leptomeningeal capillary malformations, and glaucoma [1, 5]. His incidence is not perfectly known, and estimated to be 1 in 20,000-50,000 live births, and usually has a progressive course in early childhood characterized by seizures [3-5]. SWS affect males and females equally and there is no race predilection [3, 4].

Sturge-Weber syndrome is a sporadic affection caused by somatic mosaic mutations in the GNAQ gene which is located on the long arm of chromosome 9. It has been suggested that the presence of facial angiomas is explained by the persistence of primordial sinusoidal vascular channels. Another possibility is that there is a damage of superficial cerebral venous drainage compensated by a dilatation of the cerebral veins [3, 7].

Seizures are common and often poorly managed for the patient and family. They are often bilateral in more than 90% cases, and they become progressively refractory to medication [3, 4]. Infantile spasms, cognitive impairments, increasing hemiparesis have been often associated with SWS [1, 4].

The facial nevus (wine stain) is the second clinical manifestation observed, usually involving the ophthalmic or maxillary segment of the trigeminal nerve (forehead, cheeks) [3, 6]. It is present from birth, and it is usually unilateral. The prevalence of glaucoma amongst SWS patients varies from 30% to 60% [4]. Glaucoma is often ipsilateral to facial angioma [3, 9]. Annual examinations with intraocular pressure monitoring are recommended [4].

A diffuse choroidal hemangioma visualized on the same side as a wine stain on the face is seen in about 20% of patients with Sturge-Weber syndrome [3].

Historically, plain skull X-rays and angiography help to make the diagnosis of SWS, associated to the classic "tram-line" or "tram-track" calcifications recognized in older children on X-rays, but these exams are nowadays neglected [4].

Computed tomography (CT) is the best modality to detect calcifications but it can also show other abnormalities such as cortical atrophy and leptomeningeal enhancement after contrast injection. On the other hand, it is radiating and should be avoided in children. The cause of these calcifications is attributed to chronic ischemia due to impaired venous drainage [1, 3].

Therefore, MRI of the brain with contrast is the modality recommended imaging of choice [1, 3]. The occipital and parietal/temporal lobes are the most affected sites.

MRI findings are related to the stage of the disease. In the early phase, there is leptomeningeal enhancement (seen as serpiginous enhancement along the sulci), with restricted diffusion if there is associated ischemia. In the late phase, there is a T2 hypersignal in the region of gliosis with decreased pial enhancement associated with cortical atrophy. Gyriform calcifications are best seen on T2* or SWI sequence (susceptibility weighted images) and appeared as areas with lack of signal translation along the gyri in a serpentine pattern [3, 4].

The treatment is mainly symptomatic and consists of minimizing the frequency of seizures. Surgery may be considered in patients with refractory seizures. An annual ophthalmological examination is recommended to detect ocular complications, especially glaucoma [3].

CONCLUSION

Sturge-Weber syndrome (SWS) is a neurocutaneous disorder characterized by a wine stain on the face with neurological signs. The diagnosis is provided by magnetic resonance imaging (MRI) and computed tomography (CT). The treatment is symptom based, and his management is optimal if it is multidisciplinary.

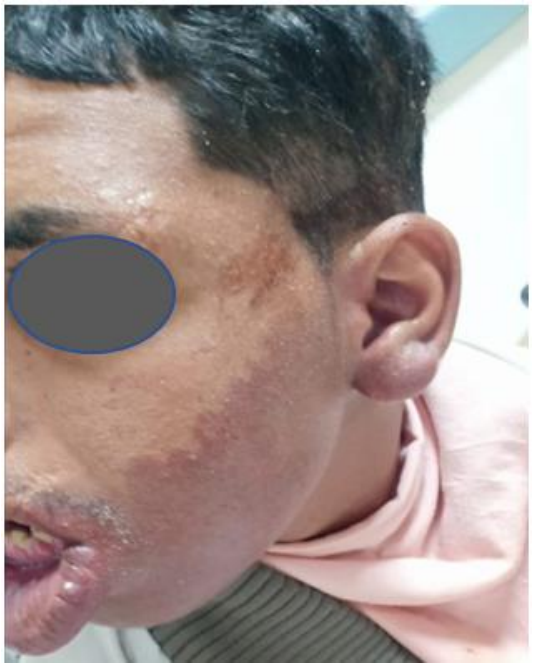

Fig-1: Cutaneous planar angioma along the mandibular segment of the trigeminal nerve

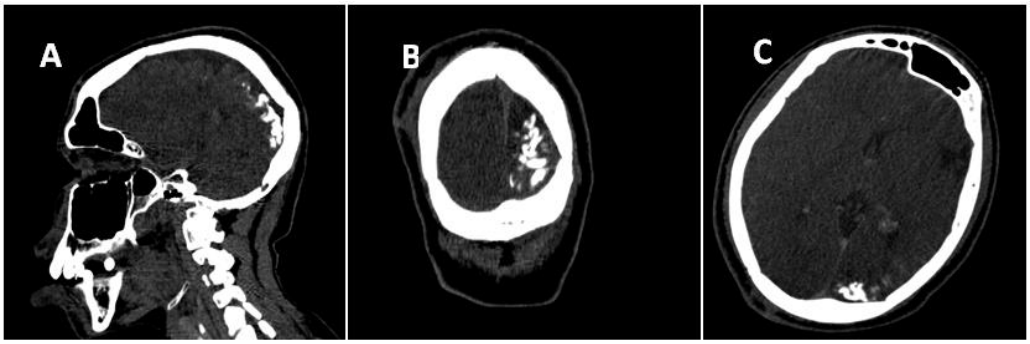

Fig-2: Representative images from Computed imaging with Sturge–Weber syndrome. Sagittal (A) and coronal (B) showing gyriform calcification. (C) postcontrast axial CT showing an enlarged choroid plexus with leptomeningeal enhancement over the left cortex.

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