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A Rare Case Report of Sturge Weber Syndrome

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Abstract: ***A 55 year old male patient with left handedness came to OPD with cause of abdominal pain, aggravating on taking food for one week having neurological signs of deviation of angle of mouth to left side, weakness in right upper limb and lower limb, recurrent episodes of seizures, hyperpigmentation on left upper half of face since childhood which was red and erythematous in childhood without involving sensory, autonomic, cerebellum, bowel and bladder control suggestive of neurocutaneous syndrome presenting with gastritis.***

Keywords: Sturge Weber Syndrome, abdominal pain

1. Introduction

In 1860, Schirmer was the first person to describe the Sturge Weber syndrome (SWS). In 1879, Sturge provided a more precise description of the syndrome, linking the dermatological and ocular alterations of the disease to its cerebral symptoms. Weber added to it in 1929 with the reporting of radiologic abnormalities detected in these individuals [1]. There are around one in fifty thousand people who are born with Sturge - Weber syndrome, making it a very uncommon condition.

The Sturge–Weber syndrome, often known as SWS, is classified under the umbrella term of phakomatoses, which literally translates to "mother - spot illnesses. " It is characterised by the combination of venous angiomas of leptomeninges, face, jaws, and oral soft tissues, and it is made up of congenital hamartomatous malformations that can affect the eye, skin, and central nervous system (CNS) at different times. In addition, it can affect the CNS at different times [2]. Typically, only one side of the body is affected by these alterations, and there is no racial disparity in terms of who displays them [3].

It is thought that SWS is brought on by the continuation of the vascular plexus surrounding the cephalic section of the neural tube after birth. This plexus begins to form during the sixth week of the formation of the I. U., although it generally disappears around the ninth week of development. Angiomas of the leptomeninges are often situated in the parietal and occipital regions and develop unilaterally. The presence of angioma causes a change in the vascular dynamics of the surrounding tissue, which leads to the precipitation of calcium deposits in the cerebral cortex underneath the angioma. A subsequent consequence of this might be the development of seizures, mental retardation, hemiplegia, or hemiparesis; the severity of these conditions is proportional to the amount of the injury. [4]

Port wine stains are cutaneous angiomas that often appear unilaterally along dermatomes that are supplied by the ophthalmic and maxillary divisions of the trigeminal nerve. These cutaneous angiomas are also known as port wine stains. It might affect both sides of the body, be completely absent, or spread to the neck, arms, and other areas of the body. Involvement of the region cared for by the ocular division is diagnostic of the condition. Glaucoma, choroidal hemangioma, bupthalmos, or hemianopis are all potential complications that might arise from ocular involvement. [5]

This condition is characterised by a group of symptoms and signs, some of which include mental impairment, focal seizures, hemiparesis, intracranial calcification, and facial nevus. Angiomas involve the leptomeninges and skin of the face, typically in the ophthalmic and maxillary distributions of the trigeminal nerve, which is why the syndrome is also known as encephalofacialangiomatosis or encephalotrigeminal angiomatosis. Both of these terms are used as synonyms for the syndrome. Oral symptoms of the illness can vary significantly, and researchers have documented alterations in the morphology and histology of gingiva, periodontium, and pulp as potential outcomes. However, the gingival hemangiomatous lesion is the most prevalent manifestation of this condition [6]. This lesion is often confined to the ipsilateral maxilla, mandible, floor of the mouth, lips, cheeks, palate, and tongue. When both chromosomes are affected by angiomas, development problems are more likely to occur [7].

2. Case Report

A case of 55 year old male patient with left handedness came to OPD with cause of abdominal pain, aggravating on taking food for one week having neurological signs of deviation of angle of mouth to left side, weakness in Rt. UL &LL, recurrent episode of seizures, hyper pigmentation on left upper half of face since childhood which was red and erythematous in childhood without involving sensory, autonomic, cerebellum, bowel & bladder control suggestive of neurocutaneous syndrome, presenting with gastritis.

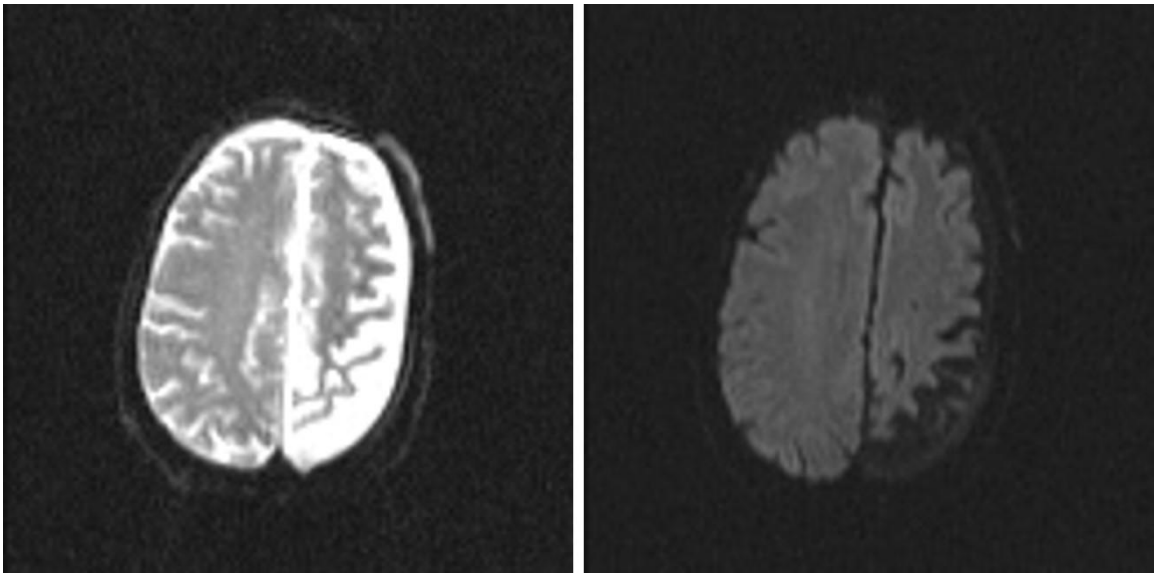

Figure 1: Showing Unilateral left sided atrophy of cerebral hemisphere with gyriformcortico - subcortical calcification of lobes and mild thickening of overlying calvarium

The patient was admitted to the Department of General Medicine. On investigations the MRI brain scan revealed gyral calcifications and severe atrophy in the right cerebral hemisphere (Fig.1). There was a significant compensatory subdural fluid collection with diffuse gyral calcification in the cortex. This study is aimed at documenting a rare case of SWS incidentally.

3. Discussion

When both CNS and face angiomas are present, a diagnosis of SWS is considered to be full. However, when only one of these areas is afflicted, the diagnosis is considered to be partial. Classification is done with the use of the Roach scale [7].

1) Angiomas of the face and the leptomeninges are present in type I - Consists of skin and neurological symptoms. These individuals may or may not have glaucoma.
2) Angiomatosis of the face only in Type II - Consist of skin symptoms and possibly glaucoma, but there is no evidencevof neurological involvement.
3) Isolated leptomeningeal angiomas; often no glaucoma is present in patients with type III - Consists of neurological involvement, but without skin abnormalities. Glaucoma is usually not present. Type 3 may also be known as the isolated neurological varient

The neuroimaging techniques allow for an accurate assessment of both the diagnosis and the management of the condition. Previous studies have reported a variety of imaging findings of SWS, some of which include atrophy of the ipsilateral hemisphere, enlargement of the ipsilateral choroid plexus, leptomeningeal enhancement, thickened calvarium, expansion of the paranasal sinuses and mastoid air cells, and white matter change [4]. The MRI scan in this instance indicated that the patient had unilateral left sided atrophy of the cerebral hemisphere along with gyriformcortico - subcortical (tram track) calcification encompassing all lobes. On the other hand, the other observations, such as an augmentation of the leptomeninges, an expansion of the paranasal sinuses and mastoid air cells, and a thicker calvarium, were not observed in this particular instance. Previous studies found that leptomeningeal enlargement was the most specific result, whereas enlarged calvarium was more common in adult patients [4]. In the same way, an increased subarachnoid space was noticed on the left side of this patient's head, along with a little thickening of the calvarium that overlaid it. The accumulation of cerebrospinal fluid (also known as CSF), which was caused by significant brain atrophy, is one of the most notable aspects of our case.

4. Conclusion

The wide range of clinical signs associated with Sturge - Weber syndrome demonstrates both the complex nature of this condition and the challenges associated in making a diagnosis. Because the precise etiopathogenesis of the disease is unknown, effective prevention is challenging, and accurate early identification is absolutely necessary. In our case report we emphasize that rederythmatous lesion on left upper half of face during childhood and later it converted to hyperpigmented lesion. Hemi atrophy of left cerebral hemisphere and tram track calcification in favor ofSturge - Weber syndrome. For treatment of uncontrollable seizures hemisphrenectomy is indicated.

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