Sturge-Weber syndrome: Report of a case and literature review

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ABSTRACT

Sturge-Weber syndrome also called as encephalotrigeminal angiomatosis is a rare congenital neurological and skin disorder caused by persistence of transitory primordial arteriovenous connection of the fetal intracranial vasculature. It is characterized by vascular malformation with capillary venous angiomas that involve face, choroid of eye and leptomeninges with resulting neurological and orbital manifestations. The diagnosis is usually considered in a child presenting with seizures and facial capillary malformation along the trigeminal nerve distribution. The characteristic imaging features on computed tomography and magnetic resonance imaging lead to the diagnosis. We report a case presenting the atypical and radiographic features of this syndrome.

Key words: Computed Tomography, Encephalotrigeminal Angiomatosis, Leptomeninges, Magnetic Resonance Imaging, Seizures

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INTRODUCTION

Sturge-Weber syndrome (SWS) or encephalotrigeminal angiomatosis belongs to a group of disorders known as the phakomatoses (neurocutaneous syndromes involving the central nervous system [CNS]).[1] It is a congenital disorder of the vasculature of the meninges, the brain, the face, and often the eyes, and is often associated with seizures and other neurologic complications, including mental retardation, contralateral hemiparesis, and glaucoma.[2-4]

Sturge-Weber syndrome occurs sporadically with equal frequency in boys and girls.[4] It has been proposed that SWS is the result of a somatic mutation in the affected areas.[5,6] Chromosomal rearrangement and trisomies in chromosomes 4 and 10 have been detected in the affected skin of 2 patients with SWS who lacked these findings in normal skin and blood.[7]

It is believed that SWS results from a developmental defect in the 1st month of gestation, represented by the persistence of a vascular plexus around the cephalic portion of the neural tube.[8] This plexus develops in the 6th week of intrauterine life, but normally undergoes regression during the 9th week.[9] The association between cerebral angiomatosis and facial nevus, characteristic of the syndrome, is attributed to the development of the skin of the face from the ectoderm that covers this vascular plexus.[10,11]

The aim of this article is to describe the clinical case of a patient with SWS, with neurologic, ophthalmic, facial, and oral involvement.

CASE REPORT

A 12-year-old, mentally subnormal female patient reported to the Department of Oral medicine and radiology. The patient's mother gave a history of pain due to multiple carious teeth since 2-3 months. The child was the product of full term, uncomplicated pregnancy and delivery. The mother gave a history of epilepsy since 7 months of age for which the patient was given homeopathic medication. The details of the medications were not known. Still, the patient had 1-3

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epileptic attacks per day. Her mother had also noted that, since 7 months of age, she was not using her left arm and leg. She had never visited a dentist earlier. There was no family history of angiomas or epilepsy. She was totally dependent on her parents for her daily living activities.

General examination revealed hemi paresis of the left half of the body, developmental delay of motor and speech function and behavioral problems (extremely negative Frankel's behavior). There were no evidence of ocular, cardiovascular, respiratory, genitourinary or gastrointestinal diseases. Extra-oral examination showed evidence of port-wine stain (PWS) on cheeks and right side of forehead, upper and lower eyelid [Figure 1]. There was a well-delineated sessile growth in relation to the gingiva (marginal and attached) of the maxillary arch and mandibular right quadrant. The color of the hyperplastic gingiva (hemangioma) was bright red, soft in consistency and there was bleeding on probing [Figure 2]. The right halves of the hard and soft palate, vestibular and buccal mucosa were also bright red in color.

Orthopantomogram revealed grossly carious 16, 36 and 46. There was periapical radiolucency in relation to 46 [Figure 3]. Computed tomography (CT) scan of mandible showed an ill-defined enhancing soft tissue in the right buccogingival space with adjacent fat stranding [Figure 4]. The right body of mandible was slightly expanded with thickened and altered bone density. Brain sections showed gyriform calcifications in the right frontal and parieto-occipital region and atrophy of the right cerebral hemisphere [Figure 5]. Skull vault was thickened in relation to fronto-parietal bones.

Magnetic resonance imaging (MRI) brain was also advised for the patient, which revealed hemiatrophy of the right cerebral hemisphere [Figure 6]. Ill-defined hypointense areas on T1-weighted images, which turn hyperintense on fluid attenuation inversion recovery images in the right parieto-occipital region suggestive of gliotic changes, were seen. Prominence of cerebellar folia on the left cerebellar hemisphere and thinning of the corpus callosum, which was more pronounced in the posterior body and splenium was seen. The meninges were enhanced in the right temporo-parietal region.

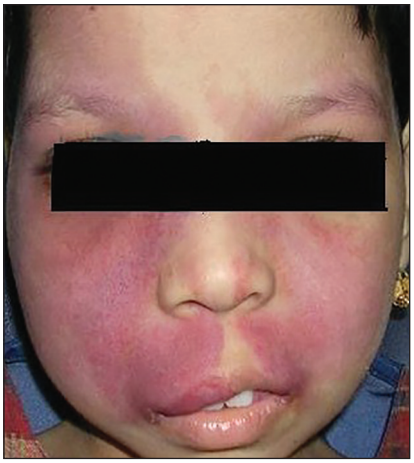

Figure 1: Bilateral facial angiomas or port-wine stains on the cheeks

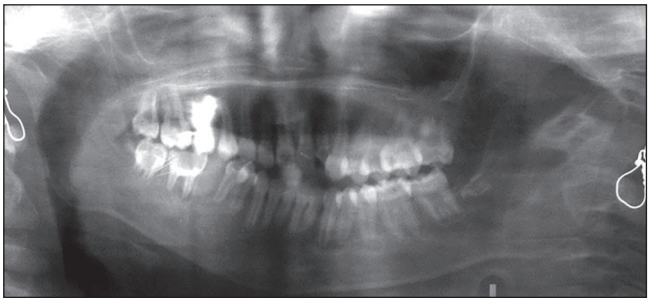

Figure 3: Orthopantomogram: reveals grossly carious 16, 36, 46 with periapical radiolucencies and lingually displaced 45

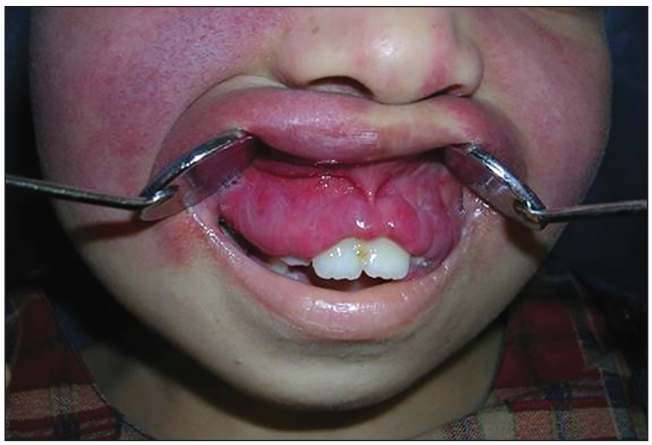

Figure 2: Well delineated sessile growth in relation to the gingiva (marginal and attached) of the maxillary arch

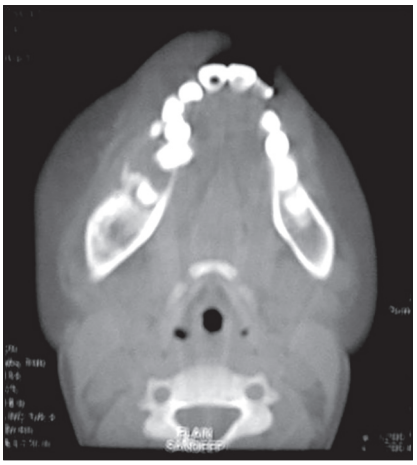

Figure 4: Computed tomography scan of mandible showing ill-defined soft tissue seen in the right buccogingival space with adjacent fat stranding

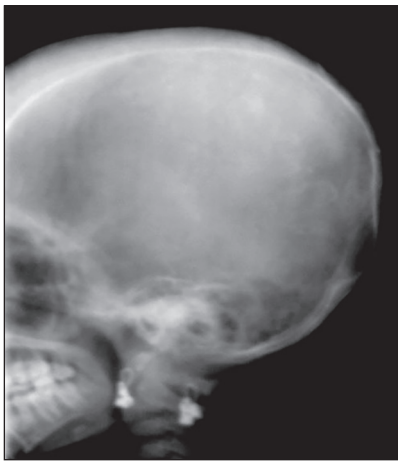

Figure 5: Skull radiograph showing gyriform calcifications

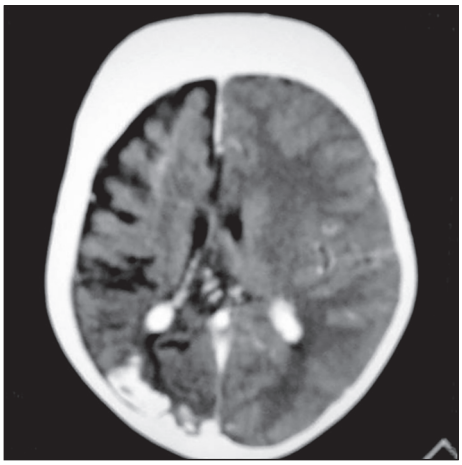

Figure 6: Computed tomography scan of the brain showing hemiatrophy of the right cerebral hemisphere. Enhancement of the meninges is seen in the right temporo-parietal region

Based on all clinical and imaging findings (PWS, hemiplegia, gingival hemangiomas, leptomeningeal angiomas) a diagnosis of SWS was given. The patient was then referred to a neurologist for management of epilepsy. The patient followed up after 4 months, when after neurological consent extractions of 16 and 46 were done under sedation. However, the parents refused treatment for gingival hemangiomas.

DISCUSSION

Clinical manifestations

Port-wine stains or nevus flammeus in SWS are well-demarcated red macular stains present at birth. Facial PWS may involve only the forehead and upper eyelid following the path of the ophthalmic branch of the trigeminal nerve (20%). Concomitant involvement of the maxillary or maxillary and mandibular branch occurs in many patients (2-23%). Bilateral lesions can be seen in 10-30% of patients.[12]

Seizures are the most common neurologic manifestations and have been reported to occur in 23-83% of patients with SWS.[12,13] In the majority of patients, they develop before 2 years of age.[13] Seizures occur either on the contralateral side of the PWS or are generalized. Additional neurologic manifestations include headache, transient stroke like neurologic deficits, behavioral problems and contralateral hemiparesis, hemiplegia, and hemianopsia.[12,13]

Glaucoma and choroidal hemangioma are the most common ocular manifestations.[14] Both the conditions are usually ipsilateral to the facial PWS. Increases in the episcleral venous pressure and developmental anomalies in the anterior chamber angle have been considered to be the main causal factors for the glaucoma associated with SWS. Choroidal hemangioma is usually seen as a red, flat to moderately elevated mass producing a classic "tomato ketchup" appearance on fundoscopic examination.[12] Other ocular abnormalities that have been reported in SWS include dilatation and tortuosity of conjunctival and episcleral vessels, buphthalmos, iris heterochromia, optic disc coloboma and cataract.[12]

Oral manifestations of SWS result from involvement of the oral mucosa by hemangioma. The gingiva of the maxilla and mandible, floor of the mouth, lips, cheeks, palate, and tongue ipsilaterally to the PWS exhibit hypertrophy and red purplish color.[15-18] Hard tissue involvement consists of bone overgrowth and tooth maleruption. Macroglossia and hypertrophy of the maxillary bone, found in some patients, might result in malocclusion and facial asymmetry. The patients may experience gingival hyperplasia secondary to pharmacotherapy for epilepsy that may be further exacerbated by poor oral hygiene associated with mental retardation.[18]

Sturge-Weber syndrome is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other. The Roach Scale[19] is used for classification. Type I presents facial and leptomeningeal angiomas and glaucoma. Intercranial angioma can be confirmed by histologic examination or typical radiographic findings. Epileptic convulsions or encephalographic alterations allow a presumptive diagnosis in the child with typical nevus. Type II exhibits facial angioma and glaucoma, without evidence of intercranial disease. Type III, which is rare, is characterized by isolated leptomengingeal angioma and absence of glaucoma.[20]

Our patient had PWS bilaterally involving ophthalmic and maxillary divisions of the trigeminal nerve. The brain however had the involvement of the right side where both

atrophy and leptomeningeal angiomas were evident. The child had a history of multiple seizures per day. Gingival hyperplasia was evident in the patient, which was not secondary to antiepileptic medications. Thus, the patient had SWS type I.

Imaging

Skull radiographs demonstrate a typical pattern of calcification resembling a "railroad track." Other indirect signs that may be seen include cranial asymmetry, thickening of the cranial diploe and increased sinus sizes on the side of the PWS.[12,21] Orthopantomograms may show unerupted teeth and osteohypertrophy of the involved side as seen in our case.

On CT scans, homolateral cortical atrophy may be seen, which may be associated with a compensatory hypertrophy of the frontal bone and frontal sinus. Cortical and intracranial calcifications underneath the leptomeningeal angiomas are best seen on CT scans. They have a characteristic gyriform or "s" shape. They may we found at the cortex as well as at the meningeal arteries, cortical, and subcortical veins. They are commonly associated with cortical atrophy in the same area. Atrophy and calcifications are considered to be an indirect consequence of chronic ischemia of the cortex due to vascular stasis in the area of leptomeningeal angioma. An ipsilateral prominent choroid plexus may be seen on contrast-CT, with or without calcifications. Sometimes this is the only sign of SWS on CT scans.

Magnetic resonance imaging without contrast may show cerebral atrophy both in T1- and T2-weighted sequences, diploic prominence, and enlarged choroid plexus. Calcifications may be seen in T2-weighted, especially, in gradient-echo images, as areas of low signal.[22,23] Accelerated myelination may produce the areas of hypointensity in T2-weighted. Hyperintensity on T2-weighted representing areas of gliosis may also be seen. MRI after contrast administration permitted a better evaluation of the extent and patency of the leptomeningeal angiomatous malformation, size of choroids plexus and the parenchymal venous anomalies.

Functional neuroimaging studies of glucose metabolism by position emission tomography (PET) or cerebral perfusion by (single photon emission computed tomography), show a transient hypermetabolism (PET) in the affected cortex secondary to hyperperfusion. At advanced stages there is hypometabolism and hypoperfusion.

Treatment

Control of epilepsy is the major goal in treatment of SWS patients. Medical treatment with carbamazepine, sodium valproate, phenobarbitol, or phenytoin have all been tried.[12] Vigabatrin may also be useful. Facial PWS can be treated with pulsed dye laser. Control of glaucoma may be achieved with medical treatment alone (beta-blockers and carbonic anhydrase inhibitors) surgical procedures have been performed, including cyclocryotherapy, YAG laser goniotomy, surgical goniotomy, and trabeculotomy or trabeculectomy.[12]

The patients with SWS should be treated when epileptic attacks are infrequent. A mouth should be used whenever possible and the oral cavity should be kept as free as possible of debris. Treatment under local anesthesia is safe, but over-enthusiastic use of lidocaine should be avoided.[24] Conscious sedation can be used when required.[24] Whenever oral surgery is planned in SWS patients, particular care must be paid to control hemorrhage during and after the surgical procedure. Postsurgical bleeding can be controlled by pressure provided by splints,[18] injection of anesthetic solutions with vasoconstrictors in the surgical sites and presurgical evaluation of hypertrophic mass vascularity by angiography.[25,26] When the extent of the lesion is judged at risk for intra and postsurgical hemorrhage (mainly when tooth extraction is planned), preoperative vascular embolization can also be used.[27] Nd:YAG and CO_2 lasers can be used for gingivectomies in SWS patients with good safety.[28]

CONCLUSION

The findings of SWS on CT and magnetic resonance include superficial cerebral calcification, cerebral atrophy, hypertrophy of choroid plexus, and leptomeningeal enhancement. Patients have neurological, ophthalmic, facial and oral involvement. Dental treatment must be planned carefully to avoid compromising the patient's health.

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