Case Report

Sturge-Weber syndrome: Continued vigilance is needed

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ABSTRACT

Sturge-Weber syndrome (SWS) is a non-hereditary congenital disorder due to somatic mosaic mutations in the GNAQ gene. The classical presentation relates to the brain lesion (cerebral angiomatous lesion of leptomeninges, which is responsible for epileptic seizures, hemiparesis and mental retardation), skin lesion (unilateral facial nevus), ocular and oral involvement. We present a 12-year-old boy who was referred to the Division of Pediatric Neurology, King Saud University Medical City, Riyadh, Saudi Arabia with left-sided hemiparesis. Physical examination showed a port wine stain involving the right side of the face, extending to the upper thorax, and enlargement of both the right eye globe and cornea (megalocornea), indicating the presence of glaucoma. Following urgent referral to ophthalmology service, his eye condition improved dramatically post surgery. Neuroradiological investigations, including cranial computed tomography (CT) and magnetic resonance angiography (MRI) revealed the classical brain lesions of SWS, as well as right leptomeningeal choroidal angioma. Ten months later, he developed focal-onset seizures which responded to treatment. His cognition is normal with good school performance. Continued vigilance is needed to identify and manage the complications of SWS.

Key words:

Sturge-Weber syndrome; Cerebral angiomatosis; Dyke-Davidoff-Masson syndrome; Cerebral calcification; Choroidal haemangioma; Glaucoma; Dental manifestations

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INTRODUCTION

Sturge-Weber Syndrome (SWS) is a sporadic neurocutaneous disease due to somatic mosaic mutations in the GNAQ gene [1]. The condition is characterized by facial port wine stains, brain lesions (leptomeningeal angiomas) and ocular abnormalities (choroidal angiomas and glaucoma) [2,3]. These are usually reflected on the patient's clinical presentation with epilepsy, hemiparesis and mental retardation as well as visual disturbances at different ages. The ocular manifestations are due to vascular malformations involving conjunctiva, sclera, choroid and retina [4]. We report here a child with SWS who was referred because of left-sided hemiparesis. Physical examination revealed signs of glaucoma of the right eye, which was treated surgically. Ten months later, he developed focal-onset seizures which responded to treatment.

CASE REPORT

A 12-year-old boy was referred to the Division of Pediatric Neurology, King Saud University Medical City, Riyadh, Saudi Arabia with left hemiparesis and right-sided facial nevus since birth. He was born at term to consanguineous parents by spontaneous vaginal delivery without postnatal complications. He started to walk with limping by the age of 2 years. His cognition was normal and he had good school performance. Physical examination showed a port wine stain involving the right side of the face, extending to the upper thorax, and enlargement of both the right eye globe and cornea (megalocornea), indicating the presence of glaucoma (Figure 1).

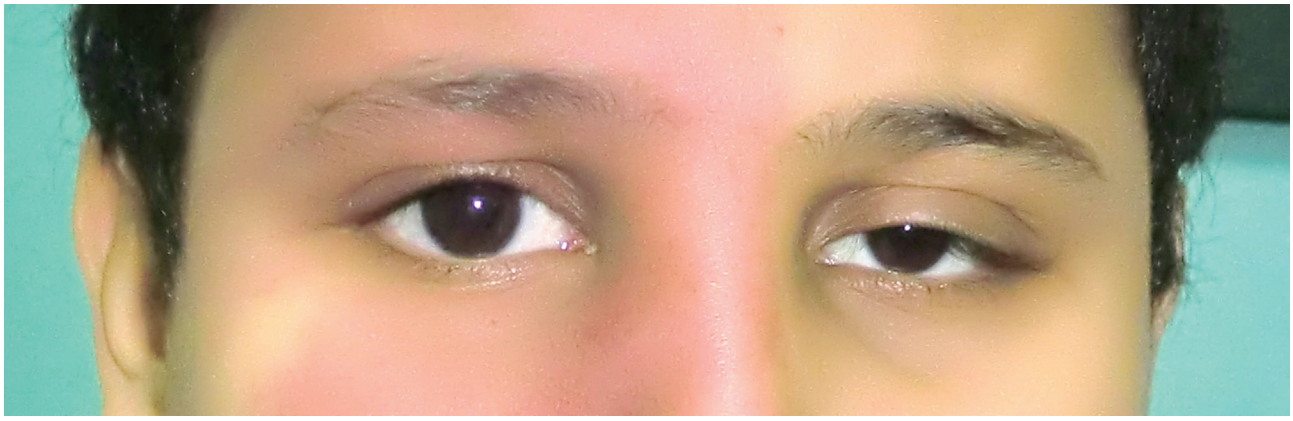

Figure 1 – Facial port wine stain and ocular involvement in 12-year- old patient with Sturge-Weber syndrome. The right eye globe and cornea are larger than the left.

He was also noticed to have gum hypertrophy. His neurological examination was significant for left-sided hemiparesis with hypertonia and hyper-reflexia in addition to a limping gait. The rest of systemic examination was unremarkable. Dental assessment revealed bilateral gingival overgrowth, poor oral hygiene with multiple dental caries and class II dental malocclusion (Figure 2).

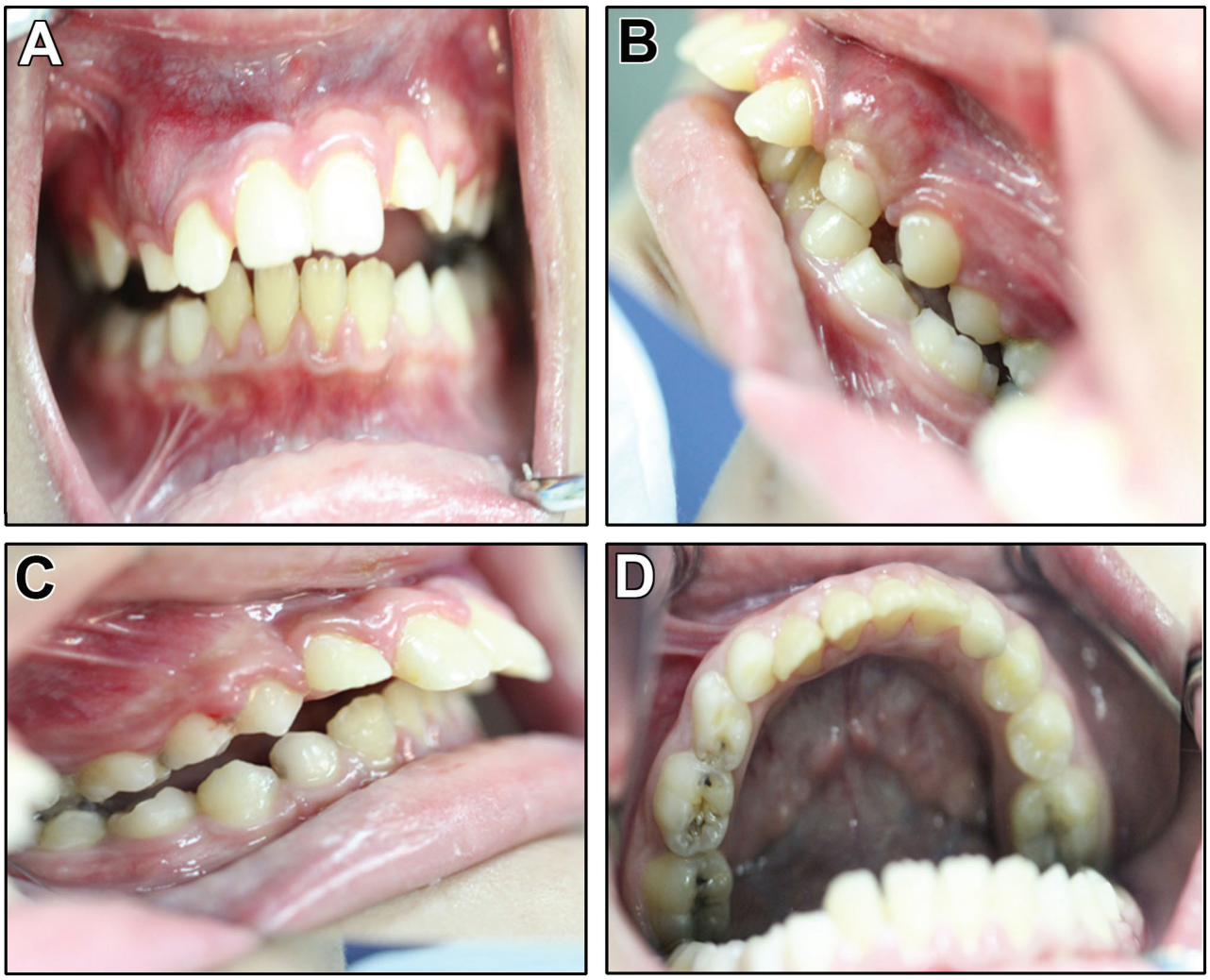

Figure 2 – Oral manifestations in the patient. (A) Gingival hypertrophy; (B and C) gingival hypertrophy and dental malocclusion; (D) haemangiomas in the mouth floor and poor oral hygiene with multiple dental caries

Examination of his right eye, following urgent ophthalmology referral, showed glaucoma with choroidal haemangioma and amblyopia. The rest of systemic examination was unremarkable. Cranial computerized tomography (CT) revealed atrophy of the right cerebral hemisphere with dense gyriform calcification (Figure 3).

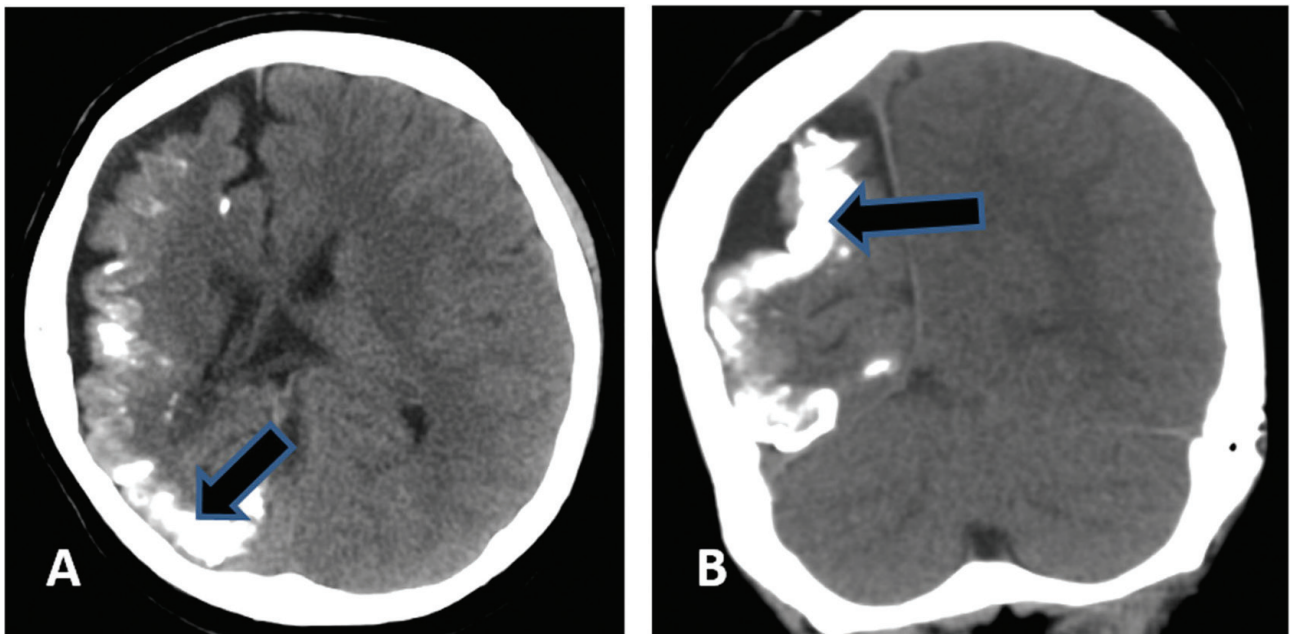

Figure 3 – Cranial computed tomography (CT) scan. Axial (A) and coronal (B) showing atrophy of the right cerebral hemisphere with dense calcification of thick cortex (arrows).

Magnetic resonance imaging (MRI) of the brain with contrast (Figure 4) showed atrophy of the right cerebral hemisphere with thick cortex, and features of Dyke-Davidoff-Masson syndrome (compensatory calvarial expansion as a consequence of long-standing cerebral hemiatrophy) [5]. There was also enlargement of the right side of frontal sinus, enhancement of the surface of the right cerebral cortex (representing pial angioma) and thick choroidal enhancement of right eye globe representing choroidal hemangioma. He received dorzolamide (carbonic anhydrase inhibitor), timolol, and latanoprost for glaucoma followed by sclerectomy with mitomycin application and gradual decompression of his right eye under general anaesthesia. His eye condition improved dramatically post surgery and he continued on maintenance medications. He was also started on physical and occupational therapy. Ten months later, he developed focal-onset seizures which responded favourably to treatment.

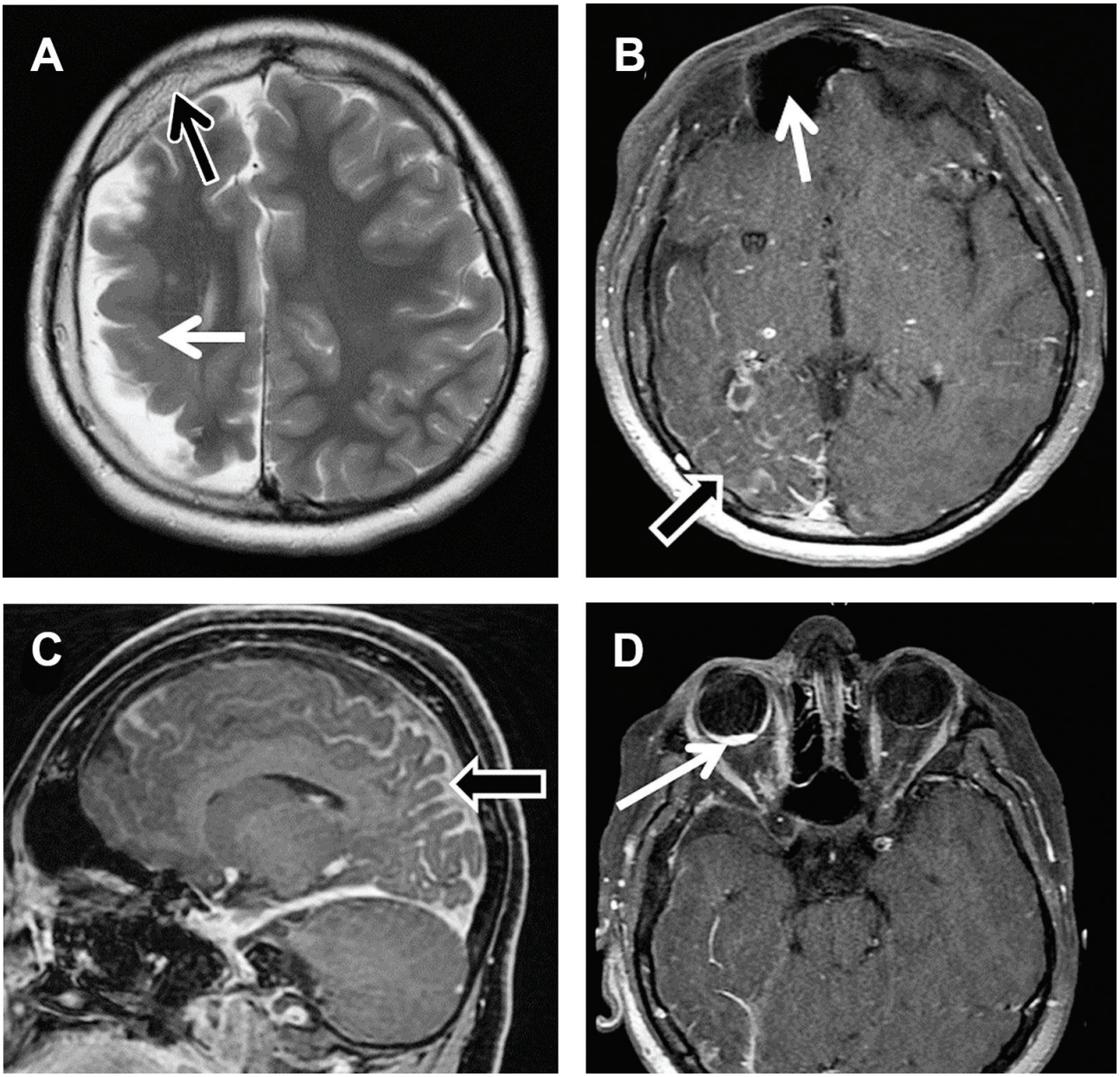

Figure 4 - Brain magnetic resonance imaging (MRI), axial T2WI (A). Axial (B) and sagittal (C) post contrast T1WI and axial orbit post contrast T1 fat sat (D). There is atrophy of right cerebral hemisphere with thick cortex (white arrow in A), thick calvarium (solid black arrow in A), large right side of frontal sinus (white arrow in B), enhancement of the surface of the right cerebral cortex representing pial angioma (open black arrow in B and C) and thick choroidal enhancement of right eye globe representing choroidal hemangioma (white arrow in D)

DISCUSSION

Sturge-Weber syndrome (SWS) is a neurocutaneous disorder of multisystem involvement.

The central nervous system (CNS) manifestations include epilepsy (75-95%), cognitive impairment and hemiparesis [2]. Hemipresis often follows the onset of seizures [6] and SWS is also considered as one of the risk factors for stroke in children [7]. The primary cerebral lesion in SWS is the leptomeningeal capillary-venous malformation (leptomeningeal angioma), which is usually located unilaterally over the posterior temporo-parietal and occipital areas, but can occur without an associated facial nevus [8]. Ischemic changes in the tissue surrounding the lesion with unilateral parietal and occipital lobe gyriform calcification lead to convulsions, hemiparesis and cognitive dysfunction [9]. In the present report, the patient had contra-lateral hemiparesis with gyriform calcifications on CT brain; however, he had normal cognition. Intellectual disability was reported to occur in 60% of SWS patients and was severe in 33% [10].

Port wine stain is the most common clinical manifestation of SWS, usually presenting unilaterally, typically on the forehead and upper eyelid, and it may be extended to the neck and other parts of the body [11]. This is similar to findings in the present case with port wine stain involving the right side of face and extending to the upper thorax. Angiomatous lesion involving oral cavity is usually unilateral and described as gingival maxillary hypertrophy leading to mal-occlusion and facial asymmetry as seen in our case [12,13].

The most common ocular presentation in SWS is glaucoma (increased intraocular pressure), which occurs in 30-70% of affected patients [14,15]. Other ocular manifestations include vascular malformations of the conjunctiva, episclera, choroid and retina [4]. About 60% of patients with SWS have glaucoma diagnosed in infancy [3] and present with enlargement of the globe and cornea, similar to the present patient (Figure 1). However, glaucoma can develop occasionally in adulthood [3]. Topical pharmacotherapy (beta-blockers and carbonic anhydrase inhibitors) may normalize the intraocular pressure, yet the majority of patients require surgical management [16].

Dental manifestations may include bleeding on probing and overgrowth in the floor of the mouth and buccal mucosa with bright red colour, which blanch under pressure [17]. The oral hygiene can be poor in SWS patients either because of recurrent bleeding, poor accessibility and/or avoidance of local trauma during brushing. Moreover, the local swelling can predispose to dental caries due to potential impaction of food particles and plaque accumulation. However, in our patient, the class II malocclusion may not be directly related to his SWS. Treatment should include advice on regular oral hygiene with regular dental visits to facilitate further intervention upon worsening of symptoms and signs.

CONCLUSION

Patients with SWS should have continued vigilance and long term follow up that includes routine workup, ophthalmology assessment, dental assessment if clinically suggested, treatment of seizures and further investigations upon the emergence of additional symptoms or signs. Early diagnosis and intervention may help in preventing, or early treating, further neurological, ocular and oral complications.

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