Case Report

An interesting case of nevus flammeus with loss of vision and hemiparesis

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Abstract

Nevus flammeus is the most common benign congenital capillary malformation, often known as a port-wine stain. Sturge–Weber syndrome (SWS) is a congenital, sporadic, nonfamilial disease characterized by intracranial and ophthalmic vascular anomalies and nevus flammeus. It usually manifests as developmental delay, learning problems, paralysis, seizures, glaucoma and attention deficit, and hyperactivity disorder. A 29-year-old male patient presented with a reddish patch over the face since birth. He was found to have hemihypertrophy of face, hemiparesis of right limbs, and low intelligence quotient. On ophthalmic examination, the patient was found to have glaucoma and only perception of light in the left eye. Computed tomography brain showed atrophy of the left cerebral hemisphere and calcifications in the left frontal, parietal and occipital regions. With these findings, he was diagnosed as SWS type I. A multidisciplinary approach was followed for patient evaluation and management. This case also highlights the irreversible sequelae of this rare phacomatosis.

Keywords: Capillary malformation, glaucoma, nevus flammeus, port-wine stain, Sturge–Weber syndrome

Introduction

Nevus flammeus (port-wine stain) is a noninvoluting type of capillary malformation, due to an embryologically defective maturation of the sympathetic fibres in the dermal vessels, which results in the loss of sympathetic control, causing vascular ectasia.[1] Sturge–Weber syndrome (SWS) is a sporadic syndrome, which is characterized by a triad of port-wine stain over the trigeminal nerve dermatome, choroidal vascular malformation of the eye and leptomeningeal angiomatosis.[2] This case emphasizes the importance of neurological and ophthalmic complications of SWS, which should be kept in mind by primary-care physicians when approaching a patient with nevus flammeus. So earlier evaluation of patient will prevent irreversible complications such as hemiparesis and blindness. SWS was first described by Schirmer and later by Sturge in 1879, while Weber described the radiological findings in 1929. SWS has no racial or sexual predilection, with an incidence of 1 per 50,000 live births.[3]

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Case Report

A 29-year-old man presented with a gradually progressive reddish discolouration of skin over the left side of the face since birth, visual impairment in the left eye since childhood and weakness of right upper and lower limbs for the past 10 years. Examination revealed an ill-defined, diffuse, barely noticeable, hyperpigmented patch extending over the left side of the face involving the forehead, cheeks, upper lip and chin. A single, skin-colored, dome-shaped nodule of approximately 5 mm diameter was noted over the left cheek. A few skin colored

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to whitish papules were present over the glabella and near the medial canthus of the left eye. Left-sided hemihypertrophy of the face was present [Figure 1]. Hemiparesis of the right upper and lower limb was noted. An erythematous patch was noted over the left buccal mucosa and hard palate, with sharp midline demarcation [Figure 2]. Other ocular findings included a red patch over bulbar conjunctiva, buphthalmos, blindness (only light perception), ciliary staphyloma [Figure 3] and glaucoma in the left eye, with an intraocular pressure of 64 mm Hg). The right eye was clinically normal. On slit-lamp examination, Haab's striae were noted in the left cornea. Biopsy of the nodule on the left cheek showed a nodular lesion in the upper dermis made up of lobules of proliferating capillaries lined by plump endothelial cells. Occasional cavernous type of blood vessels, intervening fibrosis and mild lymphoplasmacytic infiltrate were noted. Computed tomography (CT) brain revealed cortical and gyral calcifications in the left cerebral hemisphere [Figure 4]. Atrophy of left cerebral hemisphere was noted and the choroid plexus was enlarged on the same side. Widening of frontal, parietal and occipital bones with prominent diploic spaces was noted. Considering the clinical features, histological and radiological findings, we made a diagnosis of SWS. Glaucoma and hemiparesis were medically managed.

Discussion

Nevus flammeus can present separately or may be seen as a part of syndromes, such as SWS, Proteus syndrome, Parkes–Weber syndrome, Klippel–Trenaunay–Weber syndrome and arteriovenous malformations.[4] SWS, also called as encephalotrigeminal angiomatosis, is an uncommon, noninherited disorder and is due to somatic mutations in the GNAQ gene.[5,6]

Within a typical port-wine stain, small angiomatous nodules may appear, or rarely, red-purple nodules in a cobblestone pattern entirely covering the port-wine lesion may also occur. In 88% of SWS patients, the nevus flammeus involves the V2 branch of the trigeminal nerve, sometimes involving either or both V1 and V3 dermatomes. In 91% of SWS cases with eye or CNS manifestations, both eyelids were involved. In patients

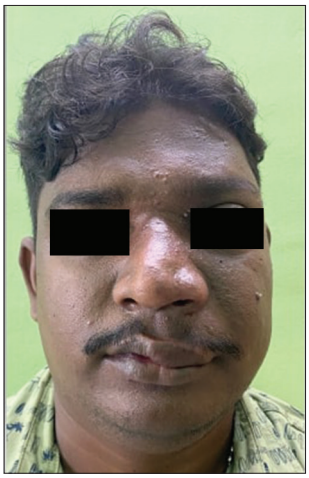

Figure 1: An ill-defined, diffuse, barely noticeable, hyperpigmented patch over the left side of the face with left sided face hemihypertrophy

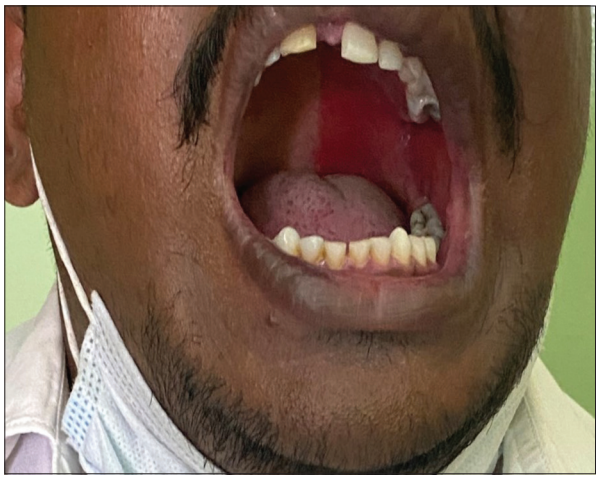

Figure 2: Erythematous patch over the left buccal mucosa and hard palate

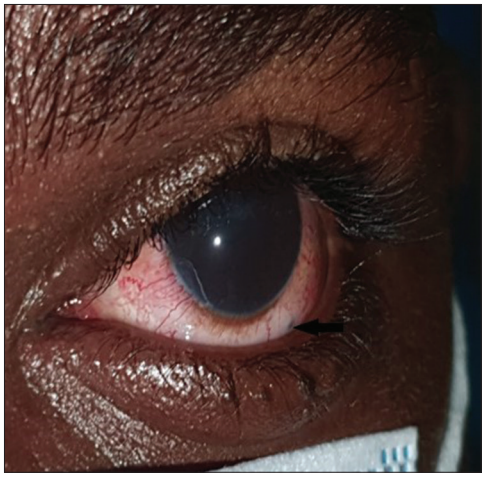

Figure 3: A red patch over the bulbar conjunctiva with ciliary staphyloma (arrow) noted in left eye

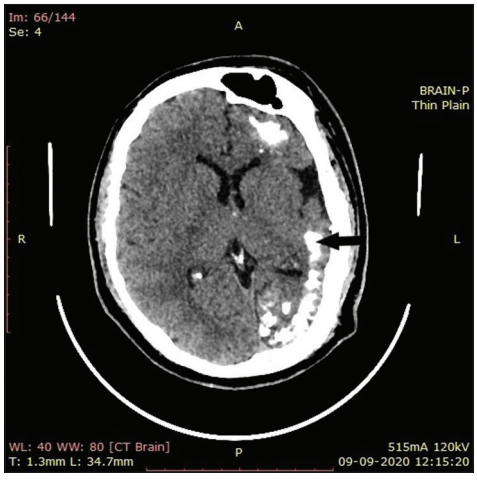

Figure 4: CT brain showed calcifications (arrow) in the left frontal, parietal and occipital lobes and atrophy of left cerebral hemisphere

with only upper eyelid involvement, no eye or CNS symptoms were noted.[7]

Neurological symptoms like seizures are caused by hypoxia and microcirculatory abnormalities. By 3 years of age, partial seizures may be seen in 70%–90% of the patients with SWS. Behavioral problems, subnormal intelligence and contralateral hemiparesis and EEG abnormalities may be seen. We observed subnormal intelligence and contralateral hemiparesis, but seizures were conspicuously absent.

Glaucoma is seen in 30%–70% of patients due to anomalies in the anterior chamber angle and increased episcleral venous pressure.[8] It causes buphthalmos, visual loss and field defects as in our case. Choroidal haemangioma with tomato ketchup fundus is the second most common ocular finding, seen in 40% of cases with SWS.

A "vascular steal phenomenon" can occur surrounding the angioma, leading to cortical ischemia, calcification and atrophy in the cerebral cortex. On the X-ray brain, "tramline" of calcifications are seen, which corresponds to the cerebral convolutions. In magnetic resonance imaging, abnormal white matter, cortical thickening, reduced convolutions and gadolinium contrast enhancement in leptomeningeal angioma may be seen.

According to Roach scale, SWS is classified into three types -

- Type I - Both leptomeningeal and facial angiomas; glaucoma may be present
- Type II – Facial angiomas alone; glaucoma may be present
- Type III – Leptomeningeal angioma; no facial angioma & glaucoma not seen.[9]

Our patient has all features seen under type I SWS.

Differential diagnoses for SWS include Maffuci's syndrome, Klippel–Trenaunay–Weber's syndrome, Beckwith–Wiedemann's syndrome and Rendu–Osler–Weber's syndrome.

The treatment varies based on the intensity or nature of the clinical features. The facial nevus flammeus may be partially treated by pulse dye laser therapy[10] or concealed with cosmetic camouflage. Treatment must be focussed on the management of severe manifestations like seizures and glaucoma, with prescription of suitable medication or timely surgical intervention, wherever appropriate. Neurosurgical procedures such as hemispherectomy or lobectomy, have been reserved as a last resort for uncontrollable neurological manifestations, but are associated with high risk of mortality. Physiotherapy can help in recovery and rehabilitation following hemiparesis.

In conclusion, our case of Type I SWS, had characteristic cutaneous lesions (port-wine stain), neurological (hemiparesis), ophthalmic involvement (glaucoma) and radiologic findings (left cerebral atrophy calcifications in the left cerebral hemisphere). This case has been presented to emphasize on the importance of identification of port wine stain by primary care physicians followed by prompt referral to specialists for further management. Early and regular screening for neurological and ocular involvement can help reduce the chances of further irreversible complications like blindness and hemiparesis, which will otherwise significantly impair the quality of life. Annual monitoring is recommended for all SWS patients and especially in children with glaucoma.

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Conflicts of interest

There are no conflicts of interest.

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