

Sturge Weber Syndrome Type I "Plus": A Case Report

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Citation

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Abstract

We present a patient with bilateral head, neck, thorax, and limbs port-wine nevus, glaucoma, epilepsy, mental retardation, short stature and abnormalities of the intracranial occipital deep veins and venous anomalies of the posterior fossa in a rural setting of the Eastern Cape, South Africa.

INTRODUCTION

Sturge-Weber Syndrome (SWS) or also called encephalotrigeminal angiomas is a congenital, non-familial disorder of unknown incidence and cause, which is characterized by a congenital port-wine nevus (facial birthmark) leptomeningeal angiomas, and glaucoma; it is commonly complicated by epilepsy and hemiparesis. Other symptoms associated with SWS can include eye and internal organ irregularities. Each case of SWS is unique and exhibits the characterizing findings to varying degrees.

The most apparent indication of SWS is a facial birthmark or "Port Wine Stain" (PWS) present at birth and typically involving at least one upper eyelid and the forehead. Much variation in the size of the stain has been reported and may be limited to one side of the face or may involve both sides. The stain, varying from light pink to deep purple, is due to an overabundance of capillaries just beneath the surface of the involved skin. In persons with dark pigmentation, the stain may be difficult to recognize. In rare instances, there is an absence of a PWS. Atypical presentations such as: intracranial venous anomalies, soft tissue hypertrophy, phakomatosis pigmentovascularis, overlapped Klippel-Trenaunay syndrome (cutaneous hemangiomas, venous varicosities and soft tissue or bone hypertrophy of the affected extremities), headache and epilepsy, and an acute life-threatening event have been reported to the medical literature.^{1,2,3,4,5,6,7,8}

SWS 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 is used for classification, as follows: Type I: This is the most common presentation with facial and leptomeningeal angiomas.

Glaucoma may be present. Ocular involvement is normally noted within the first year of life. The sclera may appear "bloodshot" as a result of the over-proliferation of blood vessels on the eye. In rare cases, the facial and brain involvement are bilateral (involving both sides of the head). Mental and physical development can be impaired to varying degrees, depending on the degree of vascular malformation throughout the brain and eye. Type 2: This type involves a facial angioma and the possibility of glaucoma, but no evidence of intracranial disease. There is no specific time-frame for the exhibition of symptoms beyond the initial recognition of the facial PWS. Throughout the life of the individual, interrelated symptoms may manifest in glaucoma, cerebral blood flow abnormalities, headaches, and various other complications. Additional research needs to be conducted on this type of SWS to determine the course of the syndrome over its natural progression. Type 3: This type of SWS is commonly noted to have a leptomeningeal angioma, with no facial involvement and usually no development of glaucoma. Commonly referred to as forme fruste, this type is identified with brain scans. It can also be confused with other diagnoses prior to a brain scan with contrasting agent.

While social stigma is lessened by the absence of PWS, the unknown natural course of the syndrome is still frustrating for parents and professionals treating the condition.

The main objective of this article is to report a case presenting clinical features of SWS type I with associated venous anomalies in the posterior fossa, being an uncommon association not previously reported.

CASE REPORT

A 14-year old female admitted at Nelson Mandela Academic Hospital (Mthatha), South Africa, presenting a history of well-controlled epilepsy on long-term phenytoin treatment, mental retardation which necessitates primary level institutionalized care, and a bilaterally swollen red face and upper limbs since birth. The patient was previously erroneously treated for a suspected allergic reaction.

On examination the patient was in a good general condition, with normal vital signs, weight: 46.7 Kg, height: 148cms. Her face was markedly red and swollen bilaterally, more so on the left. The central forehead is spared, but the blanching hyperemia extended to both upper limbs, and involves both hands, the right more markedly than the left. Hyperplastic gum was also present.

Figure 1

Figure 1: Blanching hyperemia-involving face bilaterally, with central forehead spared.



Figure 2

Figure 2: Gum hyperplasia, with abnormal vessels evident on the hard palate.



Figure 3

Figure 3: Involvement of the distal extremities.



Systemic examination, including neurological examination, was within normal limits.

The patient was admitted, after which her epileptic treatment was changed to carbamazepine 200mg per os 8 hourly because of the history of long-term phenytoin therapy and the disfiguring gum hyperplasia and because of lack of other antiepileptic drugs. Fortunately the patient remained seizure free during the rest of her 3 weeks stay in the hospital.

A CT-scan of her brain was done, which showed focal abnormal vessels in the left temporal lobe, left occipito-parietal lobe and left cerebellar hemisphere. Calcifications of some of these vessels were also seen. The ventricles were normal.

Figure 4

Figure 4: Venous anomalies on the left cerebellar hemisphere

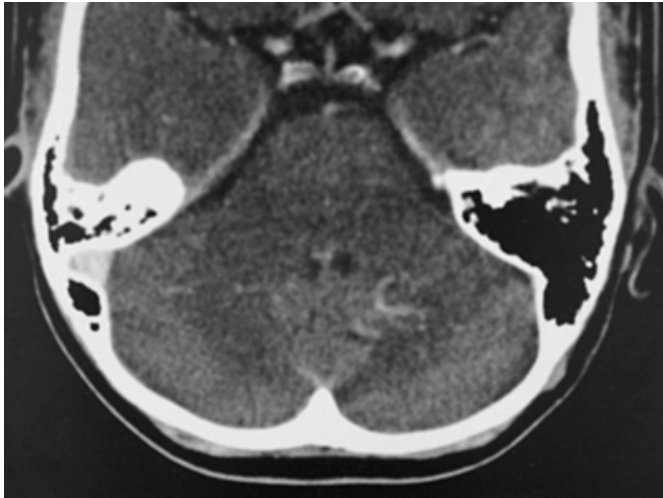


Figure 6

Figure 6: CT Scan of the brain showing angiomatosis bilateral occipital venous anomalies

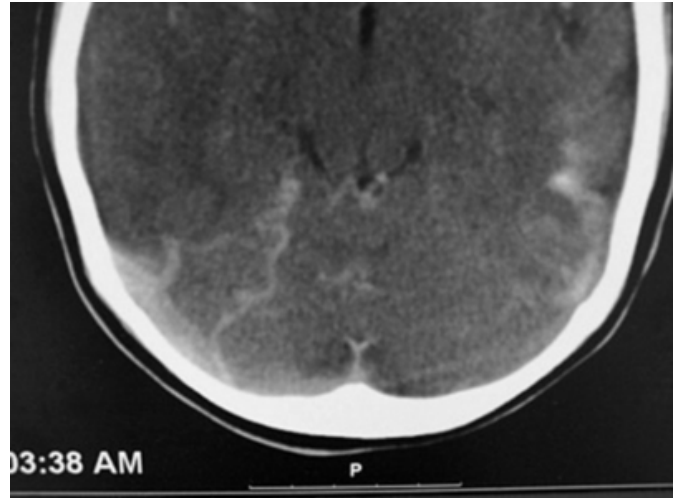


Figure 5

Figure 5: Leptomeningeal angiomatosis and vascular calcifications on the left occipital lobe.

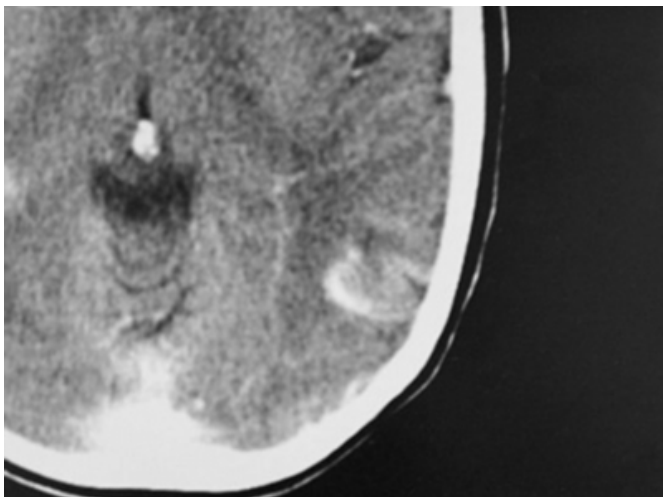
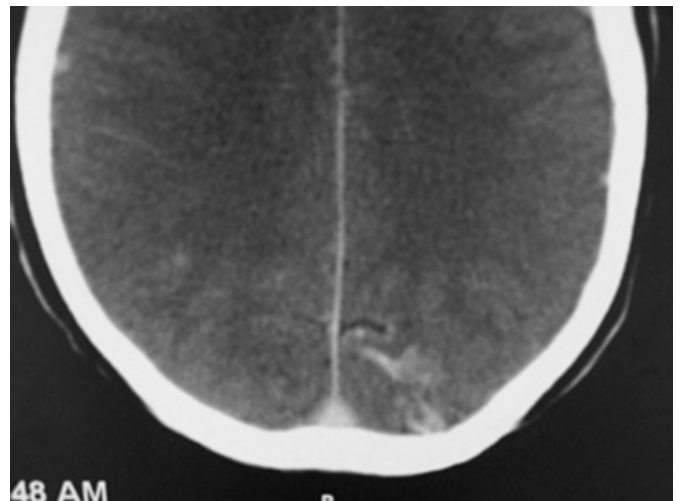


Figure 7

Figure 7: Leptomeningeal



The patient was referred for an ophthalmologic opinion. Fundoscopy through dilated pupils showed deeply cupped disks, with abnormal vessels radiating from the cups probably forming a choroidal hemangioma. The diagnosis of glaucoma was made, and the patient was started on betaxolol (beta blocking agent) eye drops, 1 drop in each eye twice a day.

COMMENTS

CAPILLARY HAEMANGIOMAS (PORT-WINE STAIN) AND STURGE-WEBER SYNDROME

Neurological concerns relate to the development of excessive blood vessel growth on the surface of the brain (angiomas). These are located typically on the occipital

region of the brain on the same side as the PWS. These angiomas create abnormal conditions for brain function in the region. Seizure activity is the most common initial presentation, often having started by one year of age. The convulsions usually appear on the opposite side of the body from the PWS and vary in severity. Vigorous attempts are made to control the seizures with medication. A weakening or loss of the use of one side of the body (hemiparesis), may develop opposite to the PWS. Developmental delay of motor and cognitive skills may also occur to varying degrees.

Capillary haemangiomas, the most common type of vascular malformation, are unsightly lesions that occur in 3 per 1000 births, with an equal prevalence between males and females. Although they are considered to be sporadic lesions, autosomal dominant inheritance has been reported⁹.

The pathogenesis of these lesions is not well understood. An array of hormonal and growth factors as well as mechanical influences have been postulated, but the primary defect that underlies haemangiogenesis remains unknown¹⁰. Recently it was suggested that haemangioma formation might be associated with somatic mutational events, with the loss of heterozygosity of a locus on 5q possibly playing a causative role in these sporadic lesions¹¹. Another study demonstrated decreased innervation in the intralesional perivascular regions, which generates the hypothesis that the lesions are secondary to impaired vascular tone⁹.

Capillary hemangiomas are commonly associated with SWS. This syndrome, first described by William Allen Sturge in 1879, with intracranial calcifications described in 1922 by Parkes Weber, is a sporadic neurocutaneous disease characterized by facial port-wine stains, ocular abnormalities (glaucoma and choroidal hemangioma) and leptomeningeal angioma^{12,13}.

Figure 8

Figure 8: William Allen Sturge, English physician, born 1850, Bristol; died 1919. For a biography click on



Figure 9

Figure 9: Frederick Parkes Weber, English physician, born May 8, 1863, London; died 1962. For a biography click on



Sturge-Weber syndrome, one of the phakomatoses, may affect the eyes, skin and central nervous system. These structures share a common embryological basis in the promesencephalic and mesencephalic neural crest, where an unknown insult results in malformations in the tissue that differentiates from it₄

Our patient falls into the diagnostic category of complete trisymptomatic Sturge-Weber syndrome, because of involvement of all three-organ systems.

Figure 10

Table 1: Clinical manifestations of Sturge-Weber Syndrome:

Risk of SWS with facial PWS *	8%
SWS without facial nevus	13%
Bilateral cerebral involvement *	15%
Seizures *	72-93%
Hemiparesis	25-56%
Hemianopia	44%
Headaches	44-62%
Developmental delay and mental retardation *	50-75%
Glaucoma *	30-71%
Choroidal hemangioma *	40%

*Manifestations found in our patient.

The ocular complications manifests as glaucoma and vascular malformations of the conjunctiva, episclera, choroid, and retina₁₆. Increased pressure within the eye (glaucoma) can be present at birth or develop later. The incidence of glaucoma in patients with SWS is approximately 70% and 40% for choroidal lesions. The glaucoma is usually restricted to the eye ipsilateral to the PWS. Glaucoma in SWS is produced by mechanical obstruction of the angle of the eye, elevated episcleral venous pressure, or hypersecretion of fluid by either the choroidal angioma or ciliary body. In juvenile glaucoma the premature aging of the trabecular meshwork of Schlemm's canal have been shown histopathologically₁₇. Decreased vision and blindness result from untreated glaucoma, with increased intra ocular pressure (IOP) leading to optic nerve damage. An acceptable range of IOP is 10-22 mm Hg₁₅. Ipsilateral enlargement of the eye (buphthalmos) can also occur.

Two disorders should be considered in the differential diagnosis of SWS₁₃:

1. Klippel-Trenaunay-Weber Syndrome: a sporadic association between capillary haemangiomas of the extremities and face, congenital venous abnormalities₁₀, hemihypertrophy of soft and bony tissues, in addition to all the characteristics of SWS.
2. Beckwith-Wiedemann Syndrome: capillary haemangioma of the face, macroglossia, omphalocele and visceral hyperplasia, with possible life-threatening hypoglycemia due to pancreatic islet-cell hyperplasia.

The focus of management in SWS is the treatment of associated neurological and ocular abnormalities¹². MRI has been reported to be superior to CT scan for detecting intracranial malformations. A- and B-scan ultrasonography and fluorescein angiography are useful investigations to diagnose a choroidal hemangioma¹⁴.

While antiepileptic treatment and the medical and surgical management of glaucoma form the mainstay of treatment, consideration should also be given to the cosmetic appearance of the disfiguring lesions. Pulsed dye lasers have become the treatment of choice in this regard, resulting in minimal scarring and preventing the natural progression to the nodular texture of lesions found in adults^{18,19}.

In patients affected by SWS some abnormalities, probably vascular related, might increase the risk of hypothalamic-pituitary dysfunction and growth hormone deficiency causing short stature²⁰. Multiple other body organs are rarely affected in SWS.

To the best of our knowledge, this is the first report on this type of association, suggesting "Type I "plus".

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