ISSN: 2474-3682

Salim et al. Clin Med Img Lib 2018, 4:092
DOI: 10.23937/2474-3682/1510092
Volume 4 | Issue 3
Open Access

Clinical Medical Image Library

IMAGE ARTICLE

Sturge-Weber Syndrome: A Diagnosis Not to be Ignored

Salma Salim*, Jamila Bouhelab and Badredine Hassam

Department of Dermatology and Venereology, Ibn Sina University Hospital, Rabat, Morocco

***Corresponding author:** *Salim Salma, Department of Dermatology and Venereology, Ibn Sina University Hospital, Rabat, Morocco, Tel: +212651705575, E-mail: salma-salim19@hotmail.fr*

Abstract

Sturge-Weber syndrome (SWS) is a neurocutaneous syndrome characterized by angiomas involving the face, choroid, and leptomeninges. The early diagnosis and the prompt treatment may reduce the incidence of neurologic sequelae, and may prevent irreversible blindness. We report a case of a 32-year-old man with SWS.

Keywords

Port wine stain, Neurological manifestations, Ocular abnormalities, Early diagnosis

Abbreviations

SWS: Sturge-Weber Syndrome; MRI: Magnetic Resonance Imaging

Introduction

Sturge-Weber syndrome (SWS) is a neurocutaneous syndrome characterized by angiomas involving the face, choroid, and leptomeninges. It is the third most common neurocutaneous syndrome after neurofibromatosis and tuberous sclerosis. It's early diagnosis and prompt treatment may reduce the incidence of neurologic sequelae, and may prevent irreversible blindness.

Case Report

We report a case of a 32-year-old man, who was treated for epilepsy for 29 years with a developmental delay and a mental retardation. Dermatological examination objectived a large and purple angiomatous lesion over the right half of the face in the distribution of the ophthalmic and maxillary divisions of the trigeminal nerve (Figure 1). Neurological examination found an hypotrophy and an functional impotence of the upper left limb which was blocked in flexion (Figure 2). A conjonctival angioma was noted in ophtalmologic examination. The MRI of the scalp revealed a cerebral atrophy. In view of the constellation of findings, a diagnosis of SWS was made.

Comments

SWS also called encephalotrigeminal angiomatosis is a sporadic and the third most common neurocutaneous syndrome after neurofibromatosis and tuberous sclerosis, and impacts approximately 1 in 20000 live births. Sturge-Weber syndrome is not inherited, but rather occurs exclusively sporadically, in both males and females and in all races and ethnic backgrounds [1]. The underlying somatic mosaic mutation causing both Sturge-Weber syndrome and isolated port-wine birthmarks was recently discovered and is an activating mutation in GNAQ [2]. It is characterized by angiomas involving the face, choroid, and leptomeninges. The facial capillary vascular malformation is also known as "port wine stain" or "nevus flammeus" and is usually found along the distribution of the ophthalmic (V1) and maxillary (V2) divisions of the trigeminal nerve. It can also involve the scalp, neck, trunk or limbs. It may remain static in extent but can undergo progressive hypertrophy, darken and become nodular in up to 65% of the patients by the fifth decade [3]. When a newborn presents with a facial port-wine birthmark on the upper face, that child has a 15-50% risk of developing

Citation: Salim S, Bouhelab J, Hassam B (2018) Sturge-Weber Syndrome: A Diagnosis Not to be Ignored. Clin Med Img Lib 4:092. doi.org/10.23937/2474-3682/1510092

Received: March 30, 2018; **Accepted:** May 07, 2018; **Published:** May 09, 2018

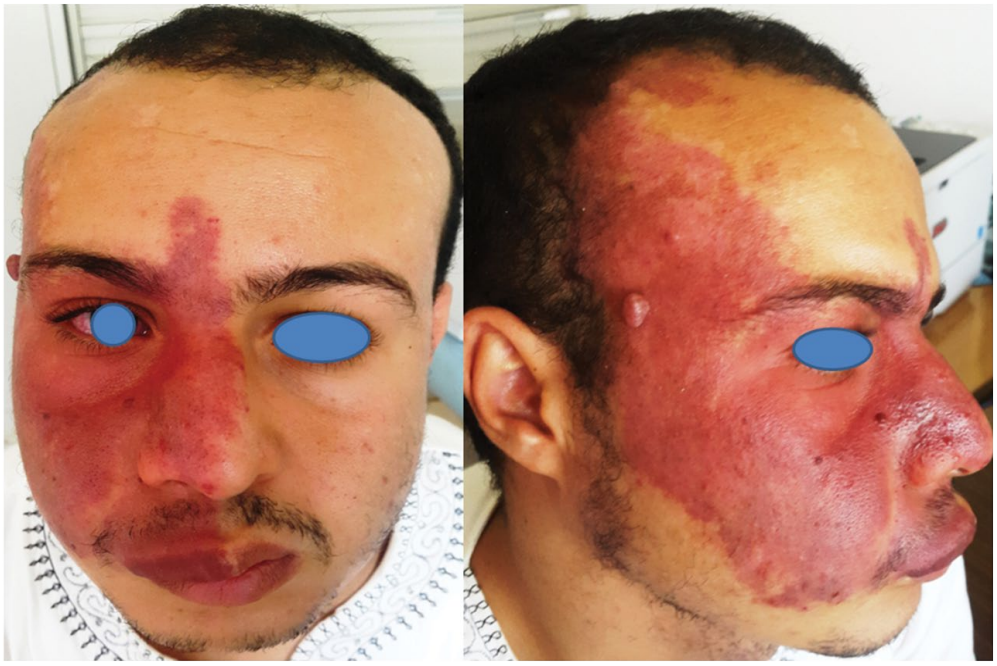

Figure 1: Port wine stain in the right half of the face (distribution in ophthalmic and maxillary divisions of the trigeminal nerve).

Figure 2: Hypotrophy and functional impotence of the upper left limb which is blocked in flexion.

Sturge-Weber syndrome [1]. A child with a facial port wine stain has 10% to 35% risk of brain involvement [4]. The leptomeningeal angiomas cause vascular steal and cortical ischemia leading to the cerebral atrophy and/or dystrophic calcifications which are best seen on MRI. Neurological manifestations include recurrent, refractory seizures (focal or generalized), transient neurological deficit, developmental delay and mental retardation. Ocular abnormalities could be choroidal, conjunctival hemangiomas and heterochromia of the iris. If there is involvement of both upper and lower eyelids, then the risk of glaucoma increases up to 50% [4]. Treatment is symptomatic with antiepileptics, antiglaucoma drugs and laser therapy for portwine stain. Low dose Aspirin has been studied in the prevention of stroke like episodes and seizures. Surgical intervention is reserved for patients with refractory seizures and uncontrolled glaucoma [5].

Conclusion

In conclusion, all patients with portwine lesions over the face, especially involving V1 region of trigeminal nerve, should be evaluated for SWS, as early diagnosis and prompt treatment may reduce the incidence of neurologic sequelae, and may prevent irreversible blindness.

Conflicts of Interest

None.

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