

## Case Report

# Sturge-weber syndrome in a three month old infant: a case report

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### Abstract

Sturge-Weber syndrome is a rare neurocutaneous syndrome that includes a facial port-wine stain and associated leptomeningeal angiomas. The defect in Sturge-Weber syndrome occurs early during lamination, when the connection between cortical veins and superior sagittal sinus is impaired. The result of which are poor cerebral metabolism, dysfunctional leptomeningeal vascular malformation, progressive atrophy and calcification. We report a case of a three months old infant who was diagnosed with clinical Sturge-Weber syndrome based on focal tonic clonic convulsions associated with weakness of the right side of her body. Physical examination revealed port wine nevi, localized on the left half of her face along the distribution of trigeminal nerve with right sided hemiparesis even though tram-line sign was not visible on her skull radiograph. Computerized tomography and MRI of the brain were desired but facilities for these were lacking at our health center. The patient was placed on Phenobarbital and diazepam for break through seizures. She improved remarkably and was discharged on physiotherapy; follow up visits in addition to seeing the ophthalmologist.

**Keywords:** Sturge-Weber syndrome, Infant, Federal Medical Centre Yola, Adamawa state, Nigeria.

## INTRODUCTION

Sturge-Weber syndrome otherwise known as encephalotrigeminal angiomas is a rare neurocutaneous congenital nonhereditary disease characterized by vascular hamartoma involving brain and facial tissues (Valbona et al., 2009, and da Conceição et al., 2011). Griffiths in 1996 documented that Sturge-Weber syndrome belongs to a category of phacomatoses characterized by nevus flammeus, angiomas of the meninges that can occur sporadically with a frequency of approximately 1/50,000 live births. Anomalous development of the primordial vascular bed in the early stages of cerebral vascularization was the theory advanced to be the cause of this disease (Erhan 2004). Erhan (2004) published that during the early stages of cerebral vascularization the blood supply to the brain, meninges and face is undergoing reorganization; while the primitive ectoderm in the region differentiates into the skin of the upper face and the occipital lobe of the cerebrum.

Both sexes are equally affected and there are no racial bias identified in the distribution of this disease.

Sturge-Weber syndrome is known to consist of a constellation of symptoms and signs including a birth mark called port wine nevi (naevus flammeus), and brain anomaly resulting from abnormal blood vessels called angiomas that are found on the cerebral cortex (Valbona et al., 2009, da Conceição et al., 2011 and Thomas-Sohl et al., 2004). Other signs and symptoms include seizures, hemiparesis, strokelike episodes, intracranial calcifications, and mental retardation. Port wine stains are usually unilateral congenital malformations involving venules, capillaries and sometimes perivenular nerves in the dermis of the skin, which often follows trigeminal nerve distribution pattern (Valbona et al., 2009, da Conceição et al., 2011 and Thomas-Sohl et al., 2004).

Valbona et al, in 2009 in Kosovo and da Conceição et al., 2011 at Rio de Janeiro Brazil have published cases of Sturge-Weber syndrome based on clinical presentation in infancy. The rationale behind diagnosing Sturge-Weber syndrome using clinical signs and symptoms at infancy was due to the fact that radiologic features appears usually from two years and above. This work reports a clinical case of Sturge-Weber syndrome in a three months-old female infant presenting with naevus flammeus, convulsions and hemiparesis that are related

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**Figure 1.** Port-wine stain (front and side view) on the left half of the face and lips

to the syndrome.

### Case presentation

A 3 months old infant presented to the Federal Medical Centre Yola (FMCY) with focal tonic clonic convulsions and weakness of the right side of her body. Physical examination revealed port wine nevi, localized on the left half of the face along the distribution of the trigeminal nerve (Figure 1) with right sided hemiparesis. Having ruled out other important differentials in our setting such as infection, trauma and bleeding diathesis; the diagnosis of clinical Sturge Weber syndrome was made based on clinical symptoms and signs. Her skull X ray did not show confluent tram-line calcifications of the left frontal bone. Computerized tomography (CT) and MRI of the brain were desired but facilities for these were lacking at FMCY. Treatment was started using Phenobarbital and diazepam for break through seizures. She responded to treatment and was discharged on physiotherapy and follow up visits. Our patient was also referred to an ophthalmologist for expert evaluation of the eye in order to check for early ocular complications, chief among these is glaucoma. Efforts were made also to educate the care-givers on the need for drug adherence and regular visits.

### DISCUSSION

Sturge-Weber syndrome has been characterized with physical, psychological, and social disorders (Valbona et al., 2009, da Conceição et al., 2011, Griffiths 1996, and

Thomas-Sohl et al., 2004). The patient is only three months old, as such it might be difficult to assess the psychological, and social components of her disease. The fact that the index patient is an infant was not surprising because Zhou et al, in 2004 reported Sturge-Weber syndrome in neonates. In fact, many authors have diagnosed Sturge-Weber syndrome in a two days old baby (Zhou et al, in 2004). Seizures occur in nearly all cases of Sturge-Weber, typically developing in the first year of life as was observed in this case report (Paller 1987). Other findings that were associated with Sturge-Weber Syndrome include cerebral calcifications, birth mark, glaucoma, hemiparesis, and cerebral atrophy. Our patient had birth mark (portwine stain) and hemiparesis that were picked on physical and neurological examinations. Sturge, in 1879 in London had argued that hemiparesis of Sturge-Weber Syndrome is caused by intracranial vessels malformations.

Radiologic findings in Sturge-Weber syndrome include cortical calcifications (tram line calcifications), cortical atrophy, enlarged ipsilateral choroid plexus, pial angiomatosis (Valbona et al., 2009, and Erhan 2004). Of note is that tram line calcifications are not usually present under the age of two years. Reason being that cortical calcifications are rarely visible in the form of tram line on skull radiograph in cases less than 2 years of age (Valbona et al., 2009, and Erhan 2004). This could explain the absence of rail road sign on the skull radiograph of our patient. Tram-line calcifications were thought to project from the left frontal sinus towards the posterior part of the left parietal region. Because the patient is only three months old and at this age sinuses are still not fully

formed; therefore, the absence of Tram-line sign may be anticipated. Better imaging modalities such as CT images for calcifications and MRI for assessing the brain in detail were desired, however, we were unable to perform these due to lack of facilities in our center.

Glaucoma is common in cases where port wine nevi are distributed in the ophthalmic and maxillary division of trigeminal nerve (Zhuo et al., 2004, Paller 1987, and Sturge 1879). The patient had port wine stain on the left ipsilateral side of her face, which also agreed with the observation made by Enjolras et al, in 1985. Port wine stains were observed to be common in cases of Sturge-Weber syndromes in children (Enjolras et al, 1985). Buphthalmos if present may be a tale tale sign of glaucoma; however, this was not seen in the index patient. Nonetheless, she was referred to the ophthalmologist on this basis. Sturge-Weber syndromes are not life threatening in most cases; however, it could be progressive associated with neurological deficit (Erba 1990). Quality management of convulsions, visual problems and hemiparesis can preserve life. The index patient was commenced on Phenobarbital and physiotherapy for the ipsilateral paralysis of the right side of the body.

## CONCLUSION

Managing Sturge-Weber syndrome demands adequate knowledge of its signs, symptoms and drug therapy. In this case report, prompt and early intervention in addition to multidisciplinary team approach was found to be effective.

## Consent

Parental informed consent was obtained for publication of this case report.

## Competing interests

None declared

## Authors' contributions

BUA reviewed, examined, treated and performed follow up of the patient. BUA, AG, collected the data, and analyzed them. All authors analyzed the image, and assisted in writing the text. All authors read and approved the final manuscript.

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