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Case Report

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STURGE-WEBER-SYNDROME IN A 23 YEAR OLD SECONDARY SCHOOL DROP-OUT: A CASE REPORT

[1]*Balarabe S.A., [2]Abubukar M., [1]Jamila M., [1]Akintomide T. and [1]Aliyu A.

[1]Department of Medicine, Usmanu Danfodiyo University Teaching Hospital Sokoto, Nigeria.
[2]Department of Surgery, Usmanu Danfodiyo University Teaching Hospital Sokoto, Nigeria.

***Corresponding Author: Dr. Balarabe S.A.**

Department of Medicine, Usmanu Danfodiyo University Teaching Hospital Sokoto, Nigeria.

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ABSTRACT

Sturge-Weber syndrome is a rare sporadic developmental neurocutaneous disorder, of undetermined etiology, that is characterized by unilateral or bilateral port-wine stain (PWS), focal or generalized epileptic seizures, hemiparesis, learning difficulties, mental retardation and ocular features such as glaucoma. It is a rare disorder occurring with a frequency of 1:50,000 live births, there is no racial or sex predilection as both sexes and races are affected equally. Currently, there are clear differences for the minimum diagnostic criteria required for the diagnosis of Sturch-Weber Syndrome, that includes a triad of cutaneous (facial) angioma, leptomeningeal angiomatosis and ocular involvement, or facial angioma and the possibility of glaucoma but with no clear evidence of intracranial disease, or just based on the presence of intracranial leptomeningeal vascular malformation. We present herein, a case of 23 year old secondary school drop-out, who presented with typical clinical and radiological features of Sturge-Weber Syndrome. To the best of our knowledge, this is the first reported case of Sturge-Weber syndrome from North-Western Nigeria.

KEYWORDS: Sturch-Weber Syndrome, Neurocuteneous disease, Epilepsy, Leptomenningial angiomatosis, Port-wine stain.

INTRODUCTION

Sturge Weber syndrome (SWS) was first described by Schirmer in 1860, later in 1879 Sturge established the relationship between skin and eye changes seen in SWS with its neurologic manifestations(Neto FXP. et al. 2008). Otto Kalischer, a German pathologist, described post-mortem findings in the disease as early as 1901, while Frederick Parkes Weber in 1929 was the first to describe the radiological appearances in this condition. Sturge-Weber syndrome is a rare sporadic developmental neurocuteneous disorder, of undetermined etiology, that is characterized by unilateral or bilateral port-wine stain (PWS), focal or generalized epileptic seizures, hemiparesis, learning difficulties, mental retardation and ocular features, such as glaucoma. It is a rare disorder occurring with a frequency of 1:50,000 live births (Welty LD. et al. 2006). There is no racial or sex predilection as both sexes and races are affected equally. (Gill NC. et al.2010).

Though the exact etiology of Sturge-Weber Syndrome is not known for certain, the disease is believed to be as a result of developmental anomaly of embryonic origin, that occur due to malformation in mesodermal and ectodermal development which in turn, results from somatic activating mutation occurring in the GNAQ gene. (Shirley, M.D.et al. 2013). The malformation is attributed to failure of regression of a vascular plexus around cephalic region of neural tube, that eventually becomes the facial skin. Failure of regression of the vascular plexus results in residual vascular tissue, which forms angiomas of leptomeninges, facial angioma and Angioma of ipsilateral eye. (Aydin A. et al. 2000). This anomaly leads to abnormal flow of blood which in turns, causes vasomotor phenomenon, resulting in ischemia, gliosis, atrophy and calcification of the cortical tissues.

Currently, there are clear differences for the minimum diagnostic criteria required for the diagnosis of Sturch-Weber Syndrome that include cutaneous (facial) angioma, leptomeningeal angiomatosis and ocular involvement, or facial angioma and the possibility of glaucoma, but with no clear evidence of intracranial disease, or just based on the presence of intracranial leptomeningeal vascular malformation. In an attempt to address the challenging issue of case definition, Roach developed a scale for classification of Sturch Weber syndrome into three types (Royle HE.et al. 1966) as follows.

Type 1: This is the most commonly described type, that manifest with both facial and leptomeningeal angiomas. Seizures usually occur in the first year of life and ocular involvement, most commonly glaucoma is common.

Type 2: This type manifests with facial angioma and the possibility of glaucoma, but with no clear evidence of intracranial disease.
Type 3: This type present with only leptomeningeal angioma, with no facial nevus or glaucoma.

Case presentation
History: We present a 23year old Hausa-Fulani, Secondary school drop-out, brought to Accident and emergency department of our hospital, with twenty two year (22year) history of recurrent focal seizure involving left side of the body. Episodes of seizure usually lasts for about 3-5minutes, mostly triggered by febrile illness. The current episode of seizure started about fifteen days prior to presentation preceded by high grade fever.

His mother noticed a red patch on the right side of his eye lid that extended to the right side of the upper lip at birth, (Port-wine stain involving maxillary branch of trigeminal nerve) which gradually regresses to the current stage of affecting only the right upper eye lid. She also noticed differential size of his limbs with the left limbs appearing smaller than the right limbs, at the age of three years, associated with weakness of the left side of the body and hemiplegic gait.

Pregnancy and purperium were uneventful, but there was history of delayed developmental milestone. Patient achieved neck control at seven months of age, was able to sit at one year, started crawling at about two years eight months and started walking at three years of age, in addition, he was only able to pronounced Mama/Baba at five years of age. There is history of difficulty in learning; he first enrolled in model Primary school at eight years of age, he takes last position throughout primary school years. He finally drop-out at junior secondary school level 3 because of poor performance.

Examination: Examination findings revealed a young man, in obvious distress, actively having focal seizures involving the left upper and lower limbs, with bupthalmos of the right eye, he was conscious but drowsy. He was noticed to have asymmetry of the skull with right more than left. Additional findings were, hemihypertrophy of the right side of the lip with hyperaemia, a small hyperpigmented nervus on the dorsum of the right hand and hyperaemic right side of upper eye lid, hypertrophy of the gum and gynecomastia. Fundoscopy revealed features of glaucomatous optic atrophy. (**Figures 1-4**).

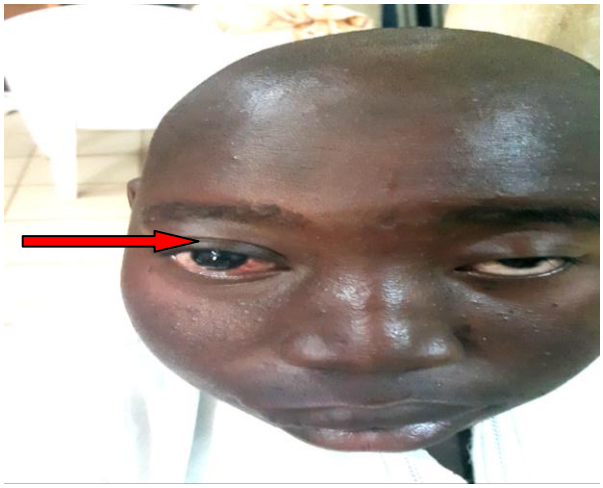

Figure 1: Showing Asymmetry of the skull with right more than left, bupthalmos of the right eye hemihypertrophy of the right side of the lip with hyperaemia and faint hyperpigmented right upper eyelid patch on the face of the patient.

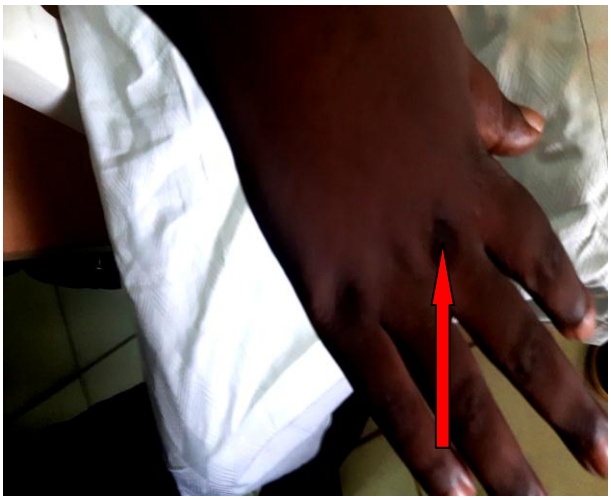

Figure 2: Showing a small hyperpigmented nervus on the dorsum of the right hand.

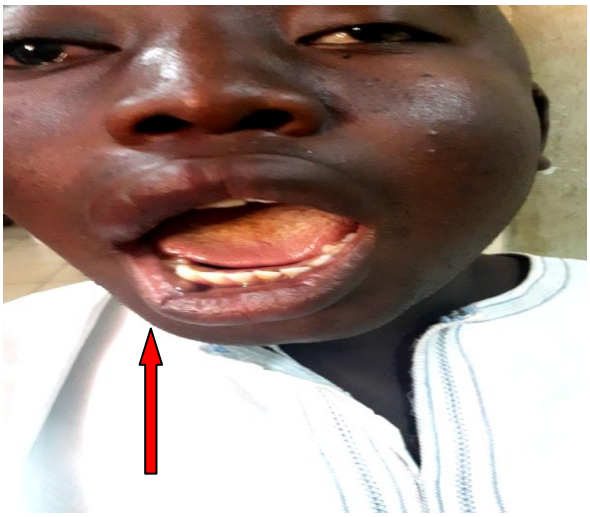

Figure 3: arrow shows right side of the lip with hyperaemia and hemangioma of the gums.

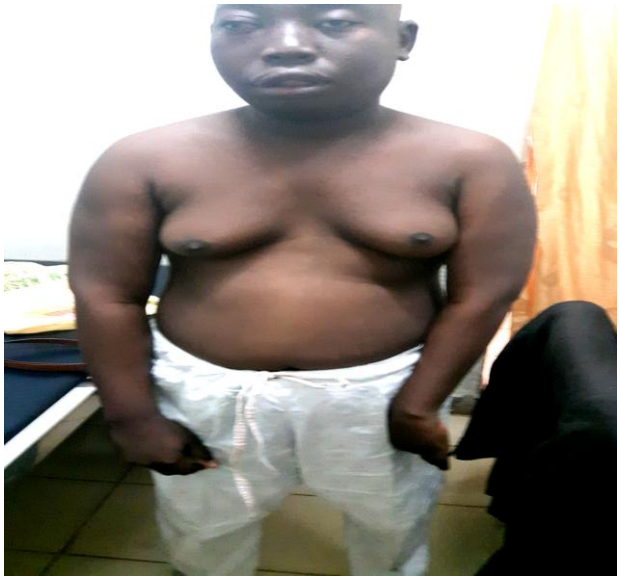

Figure 4: Showing Asymmetry of the face, disused atrophy of the left upper limb and gyneacomastia.

Skull X-Ray

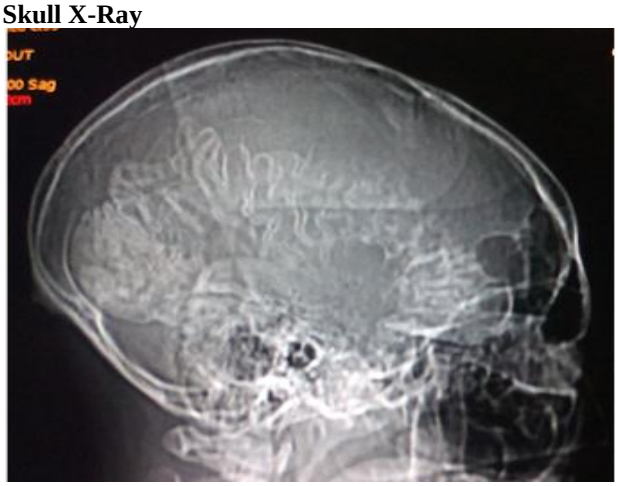

Figure 1: Plain Radiograph showing Gyriform calcification, parietal > occipital > frontal.

Computed Tomography scan: Figure A-C

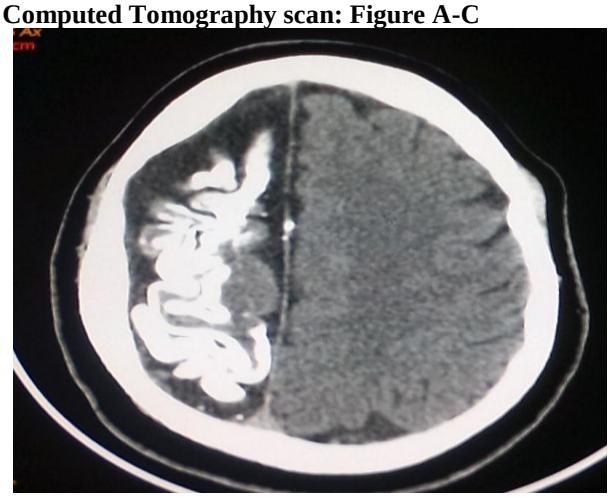

Figure 2 A: Computed Tomography scan: The sonography shows extensive coil tram track opacity of calcific density projected in the right fronto-parieto-occipital region.

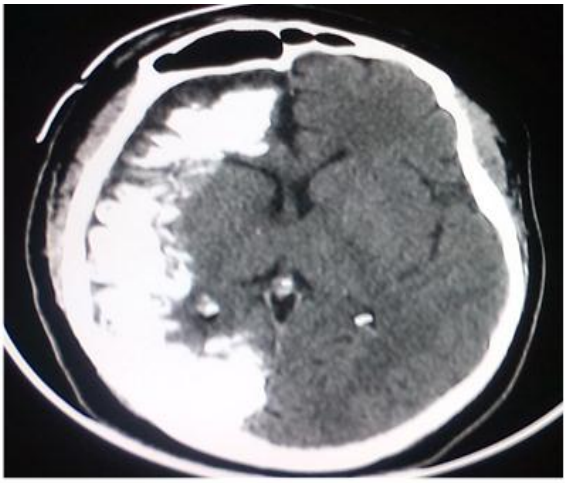

Figure 2 B: Computed Tomography scan: The sonography shows extensive coil tram track opacity of calcific density projected in the right fronto-parieto-occipital region.

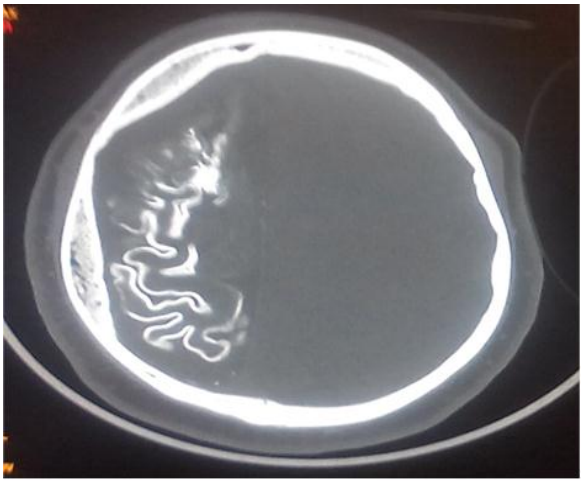

Figure 2 C: Computed Tomography scan: The sonography shows extensive coil tram track opacity of calcific density projected in the right fronto-parieto-occipital region.

Report of Computed Tomography scan: Serial axial slices done at 2.5mm interval in pre and post contrast show an extensive gyriform hyperdensity (HU=460) involving the entire right hemi-cerebrum in keeping with calcification. There is associated reduction in the right hemispheric volume. Surrounding slightly enhancing hypodensity was noted HU=5 and 8 pre and post contrast respectively. The choroid plexus in the lateral ventricles are slightly enlarged, however, the ventricular systems are not dilated. The left cerebral hemisphere shows normal density and morphology. Bone window show thickened and widen diploid space on the right side.

DISCUSSION

In Sturge Weber Syndrome, the most common clinical manifestation is childhood seizures, present in 71–89% of cases(Wyllie, E. et al. 2006). These usually begin in the first few years of life and are often associated with developmental delay and hemispheric symptoms

including hemiparesis (Muzio, B. D. and F. Gaillard. 2005-2014). The index patient developed seizure during early childhood, he also suffered delay in developmental milestone, had learning difficulty and hemipheresis. Additionally, presence of focal seizure and hemipheresis in our patient corroborated well with radiological demonstration of cerebral atrophy and evidence of calcification on the contra-lateral hemisphere.

Though, most patients with Sturge–Weber develop seizures within the first year of life, adult onset epilepsy can occur occasionally and seizure attacks are often triggered by fever. Seizure during infancy and early childhood may result in convulsive status epilepticus seen in over 50% of cases. Status epilepticus often results in a permanent and significant worsening of the neurological deficit. Our patient presented in complex partial status epilepticus.

Choroidal hemangioma causes increase secretion of aqueous humor that leads to elevation in intra ocular tension which in turn, result in buphthalmos and glaucoma(Inan C. et al. 1999) seen in about 30-70% of cases. (Anaya-Pava EJ. et al. 2015) In the present case we reported buphthalmos and glaucomatous atrophy. Skin manifestations in form of port wine stain affecting usually the distribution of the trigeminal nerve, and in about 33% of cases the naevus is bilateral. Port-wine stain may be difficult to appreciate among blacks because of their dark skin, this may account for regression of port-wine stain observed in our patient as his skin changes to typical dark skin colour because of his racial background.

CONCLUSION

Sturge-Weber syndrome is a rare sporadic developmental neurocuteneous disorder, of undetermined etiology, that is characterized by unilateral or bilateral port-wine stain (PWS), focal or generalized epileptic seizures, hemiparesis, learning difficulties, mental retardation and ocular features, such as glaucoma. Port-wine stain may be difficult to appreciate among blacks because of their dark skin, therefore, Clinicians should have low threshold for considering diagnosis of Sturge-Weber Syndrome among blacks even when they don't present with port-wine stain

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