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Case Report

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BILATERAL STURGE-WEBER SYNDROME - A CASE REPORT AND REVIEW OF LITERATURE

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CASE REPORT

A nine years old female child, the second born of a non-consanguineous marriage had multiple episodes of seizures from two years of age. Antenatal history was uneventful. The baby was delivered by Caesarean section, an indication for which was prematurity and fetal distress. The baby was admitted to the Neonatal unit for 15 days for preterm care. The baby did not have any other significant neonatal history. Family history was not significant. Delayed attainment of all the milestones was present. On examination, the child had multiple capillary haemangiomas over the bilateral upper half of the face (Fig: 1). There were no other congenital anomalies noted. The child had an intellectual disability and presented with refractory seizures. Neurological examination revealed increased tone in all four limbs and reflexes were exaggerated. CT brain showed extensive bilateral calcifications over the whole left cerebral hemisphere and right frontal lobe (Fig: 2), features of which were suggestive of Sturge Weber Syndrome. Ophthalmological examination was normal.

DISCUSSION

Sturge Weber Syndrome (SWS), also known as encephalo-trigeminalangiomatosis, is a congenital disorder with no known cause.[1] SWS is diagnosed by the triad of skin (Port wine staining), central nervous system (leptomeningeal venous angiomas) and ocular (glaucoma and choroidal haemangiomas) involvement.[1] In 1992, Roach classified SWS as follows – Type I having Neuro-Oculo-Cutaneous involvement, Type II having only Oculo-Cutaneous involvement and Type III having only Neurological involvement.[2] Bilateral port-wine staining can be seen in 10-30% of cases of SWS. The hallmark of SWS is the presence of facial nevus hammeus or port-wine stain, which is present in 96% of cases.[3] It usually occurs in the distribution of ophthalmic division of the trigeminal nerve. The present case had bilateral port-wine stain involving dermatoses of both first and second divisions of the trigeminal nerve with neurological impairment. Also, no ocular involvement was observed in this case.

SWS is a sporadic developmental disorder caused by a somatic mosaic mutation in the GNAQ gene, which is located on the long arm of chromosome 9.[4] The incidence of Sturge Weber syndrome is unknown; however, it is estimated to be one in every 20,000-50,000 live births.[5] A seizure is usually the first neurological manifestation of SWS. Seizures progressively become refractory to medication. Increased vascularity of the conjunctiva, eye enlargement, strabismus, and increased weeping are all symptoms of ocular involvement in children. The other symptoms include intellectual disability, early handedness, and gaze preferences.[6]

Diagnosis of SWS is based on typical clinical symptoms, facial appearance and brain imaging findings. Gyriform calcifications that are seen in the skull x-rays are classically referred to as “tram track sign”. A Computed tomogram is considered to be the best modality to detect the calcifications and also to find out the other changes like cortical atrophy, and leptomeningeal enhancement. There is no specific treatment for SWS. The primary aim is to control seizure activity with anticonvulsive medications. Surgery may be considered in patients who continue to have refractory seizures. The port-wine stain can be treated with laser photocoagulation which results in irreversible damage to the blood vessels without damage to the skin. An Annual ophthalmological examination is recommended to detect glaucoma.

To summarize, the lack of the classical trio of cerebral, ocular, and cutaneous dysfunction does not rule out the diagnosis of SWS. It can manifest atypically as well as bilaterally as shown in this case.

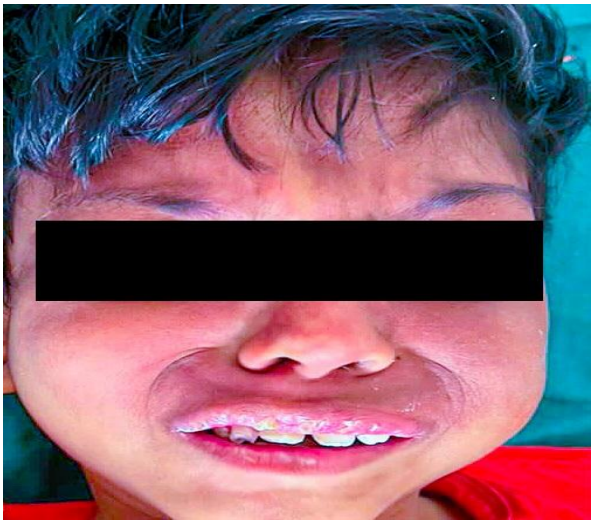

Fig. 1: Port-Wine stain seen bilaterally in upper face.

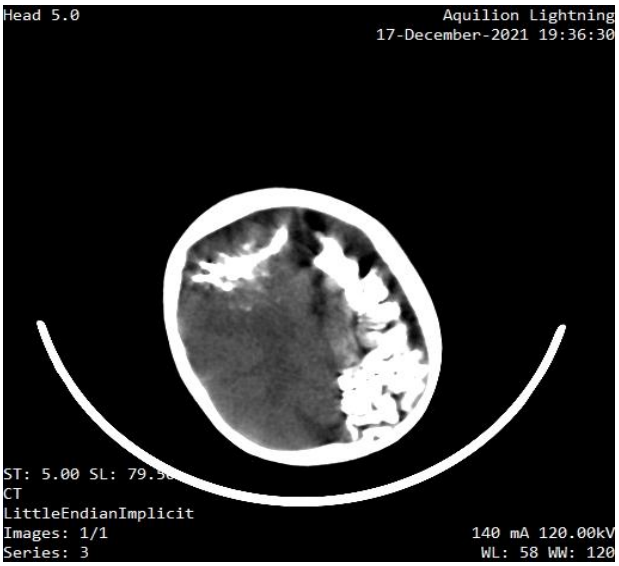

Fig. 2: CT imaging shows extensive bilateral calcifications over the whole left cerebral Hemisphere and Right frontal lobe.

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