Case Report

Sturge Weber Syndrome

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ABSTRACT

Sturge Weber syndrome (SWS) is an uncommon, sporadically occurring, frequently progressive neuro-cutaneous syndrome, consisting of congenital hamartomatous malformations, often associated with epilepsy and variable oral manifestations. The port-wine nevus (PWN) is usually the first component of the syndrome, ocular and neurological manifestations like convulsions are among the most characteristic features of this disease. Intra orally, the angiomatous lesions may involve the gingiva and buccal mucosa. Imaging modalities like conventional radiography, computed tomography and magnetic resonance imaging play a pivotal role in demonstrating the cerebral changes. Here we report a case having port-wine stain, seizures and tramline calcification in brain.

Key words : Sturge Weber syndrome, port-wine nevus, tramline calcifications.

Introduction -

Sturge Weber Syndrome is a developmental capillary vascular disorder originating during embryogenesis before neural crest migration, from errors in the development of ectoderm and mesoderm in the anterior neural primordium possibly due to somatic mutations. It is characterised by facial capillary malformations (angiomas), accompanied by variable degrees of ocular and neurological anomalies. The exact aetiology is unknown but the primary defect may be a developmental insult affecting precursors of tissues that originate in the promesencephalic and mesencephalic neural crest, which later give rise to vascular and other tissue malformations in the meninges, eye and the dermis. The orofacial manifestations are variable and include cutaneous facial portwine nevus (PWN) and ipsilateral hypervascular changes affecting the oral mucosa and gingiva. Portwine nevus is a dermal capillary vascular malformation present at birth. It is usually the first component of the syndrome to be noticed by patients. Diagnosis of SWS relies on a detailed history, thorough clinical examination and imaging studies, signifying requirement of a multidisciplinary approach. Treatment mainly consists of seizure control with anti-convulsants. Surgery is reserved for refractory seizures, intractable glaucoma and laser therapy for facial nevus.

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Case Report -

A 30 years old female patient was admitted for seizures one week back. There was no history of fever, altered sensorium, head injury or difficulty in vision. History of similar episodes of seizures was present since 2 years of age and patient was on anticonvulsant. Also there was history of right hemiparesis 5 years back which recovered completely for which she was not evaluated. There was no any significant illness in the past. General examination revealed no significant abnormality except a reddish pink nevus was present over forehead vertical, slightly left to midline since birth **(Fig. 1)**. The systemic and oral cavity examination was normal. On investigation, patient's ECG, chest X-ray, blood investigations were within normal limit. Ophthalmologic examination didn't reveal glaucoma. Patients CT head showed left sided tram-line calcification with encephalo hemangiomatosis with left sided cerebral atrophy suggestive of Sturge Weber Syndrome **(Fig. 2)**. So with typical clinical findings & CT report diagnosis of Sturge Weber Syndrome is entertained. Patient was treated conservatively and advised regular follow up.

Discussion

Sturge Weber Syndrome (SWS) also known as encephalo-trigeminal-haemangiomatosis is an uncommon congenital condition. It is a sporadically occurring, frequently progressive, congenital neuro-cutaneous syndrome often associated with epilepsy which belongs to a group of disorders collectively known as 'Phakomatoses' or 'Mother-spot disease' but in contrast to the other disorders (Neurofibromatosis, Tuberous sclerosis and von Hippel- Lindau disease), in this group, there is no evidence of heredity. The possibility of a somatic mutation being present in SWS is based on the finding of an increased gene expression of fibronectin from port-wine stain (PWS) fibroblasts when compared to fibroblasts from normal skin in the same patient. The somatic mutation in the precursors of some angiogenic factors may lead to their over production, producing angiomas or due to a lethal gene surviving by mosaicism.[1] SWS occur with a frequency of approximately 1:50,000 live births. The typical patient presents at birth with facial angiomas, however the reverse is not always true. In the incomplete form of Sturge-Weber syndrome, central nervous system angiomas occur without cutaneous manifestations, thus no suspicion of the syndrome arises until the onset of seizures.[2] Angiomas of SWS result due to failure of regression of a vascular plexus around cephalic portion of neural tube which is destined to become facial skin. This vascular plexus normally forms at the 6th week of intrauterine life and regresses by 9th week. Failure of its regression results in residual vascular tissue which forms angiomatous malformations of leptomeninges, face and ipsilateral eye.[3] There may be seizures, developmental delay and mental retardation if angiomas involve a greater part of brain. The seizures of the contralateral side to the PWS may begin in infancy and worsen with age, eventually occurring in 75% cases. Mental retardation may be progressive but is not related to the seizure severity or frequency. Patients affected with the syndrome present a variety of orofacial clinical manifestations including gingival haeman-giomatous lesions, labial angiomas and nasal septum deviation.[4] SWS is evident at birth as a unilateral PWS on the forehead and upper eyelid in the region supplied by the first branch of the trigeminal nerve, and varies in colour from pink to red to purple in colour. 50% of the affected children will have or develop glaucoma. Glaucoma may usually occur when PWS involves the eyelids.[5] The diagnosis is based on clinical and imaging studies. Skull films may reveal tram track calcification caused by calcification in apposing gyri, ipsilateral calvarial thickening and enlargement of the paranasal sinuses and mastoid. Cranial CT scan revealing cortical atrophy underlying the angioma with gyriform 'tram-track' calcifications is the characteristic imaging feature. MRI is the current gold standard for diagnosis of the disease which is reliable even in very young infants.[6] Newborn babies with a PWS should have an ophthalmological examination in the first month of life, followed by neuroimaging (CT and gadolinium enhanced MRI) by 6-12 months age or sooner if neurologic signs are present. Cerebral blood flow imaging, Single Photon Emission Computed Tomography (SPECT) and Positron Emission Tomography (PET) are also useful when possible. The neurological signs are due to ipsilateral leptomeningeal angioma involving the occipital and posterior parietal lobes of the brain; vascular stasis with resultant ischaemia leads to calcification and laminar cortical necrosis. Management of the syndrome involves both medical and surgical approaches. Medical treatment includes anticonvulsant therapy with prophylactic low dose aspirin to prevent thrombus formation. Acute treatment of seizures with benzodiazepines or if ineffective, intravenous phenytoin or phenobarbitone is recommended in India.[7] PWS may be treated with cosmetic camouflage creams, pulsed tunable dye laser and cosmetic surgery. Early surgery is advocated for better seizure control and to prevent developmental delay.[8] Behavioural problems may be encountered either due to previous exposure to hospital settings or due to mental impairment which demands the use of behavioural management techniques.

Conclusion

The Diagnosis and treatment of Sturge Weber Syndrome relies on a detailed history, thorough clinical examination and imaging, requiring a multidisciplinary approach and co-ordination between different fields of medicine, surgery and dentistry. The importance of newborn diagnosis and

further multicentre clinical investigation as well as continuing genetic research is essential in SWS. Determining the underlying cause of SWS is a difficult problem but further research in the area would allow an increase in understanding and application of new treatment options.

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Conflict of interest : Nil reported

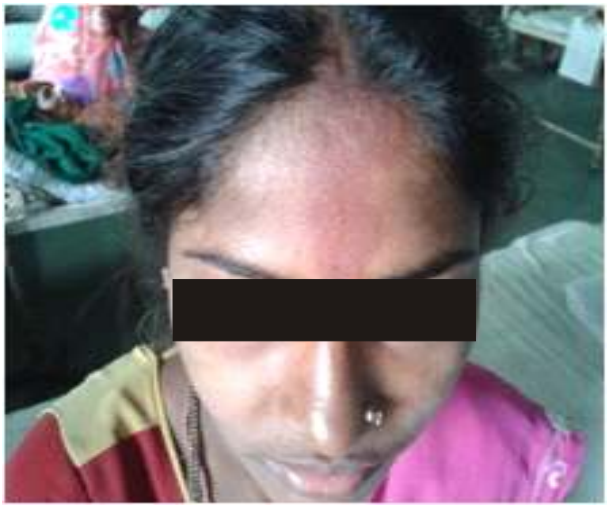

Fig 1 : **Port wine nevus over forehead**

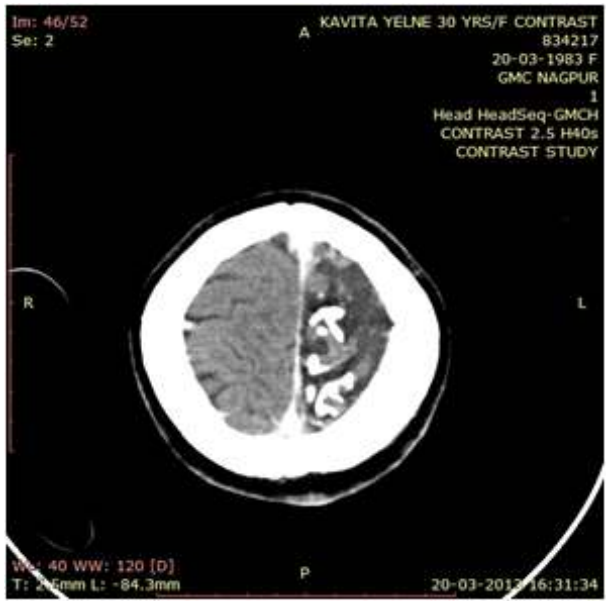

Fig 2 : **CT head contrast showing gyriform tram line calcification with left cerebral atrophy**