

Sturge-Weber-Dimitri Syndrome - An integral part of Phakomatosis

A study evaluation of 7 cases in the Department of Radiology, Mayo Hospital, Lahore

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Sturge Weber Syndrome, is a comparatively rare syndrome .The patient can present with facial manifestations, CNS manifestations , orbital anomalies and Visceral manifestations. In the last one year we encountered seven cases which were classical as far as the imaging of these lesions is concerned. These lesions were evaluated on plain film radiography and the state of art Computed Tomography, in one case the dynamic CT scan of the brain was also done. In this article we present the imaging of these seven cases .

Key words. Sturge weber, CT scan, plain film radiography

Neurocutaneous syndromes are not very common in our part of world and specially the role of imaging is not commonly known to our young doctors. This article is a presentation of seven cases which presented to the Department of Radiology with clinical suspicion of Sturge Weber Syndrome. Evaluation was done on plain films and CT scans.

Material and methods

Plain X-rays of skull AP and lateral views done on a 500mA Toshiba machine and then CT scan was performed on the X-vision Toshiba Helical CT machine. The patients ages ranged from 12 years to 44 years and all were males.

Clinical presentation.

Out of the seven cases following was the presentation of them

All seven cases presented with Seizure

Five cases with Mental deficiency

Three with Facial port wine stain

One case each with Glaucoma and Generalized weakness of body

The imaging diagnosis.

While evaluating these patients we found classic findings on the plain skull x-rays and CT scan left no doubt in the diagnosis.

Findings were as follows

1. *Skull X-Ray* Intracranial calcifications (railroad track pattern) in relation to the gyral and sulci in the occipital region.

2. *CT Scan* Leptomeningeal angiomas, atrophy of the cortex and calcifications

Discussion.

Sturge Weber Syndrome is a neurocutaneous syndrome characterized by facial nevus, ipsilateral vascular anomalies and intracranial calcifications, and contralateral hemiparesis, hemianopia, and seizures.

Epidemiologically the incidence is about 1/50000, the risk factors are familial and may be related to the chromosomal abnormalities.

Pathogenesis can be summarized as given under

- ◆ The persistence of Streeter's primordial vascular plexus
 - ◆ The primary abnormality likely arises early in development when the ectoderm which is to form the skin of the upper part of the face overlies that part of the neural tube destined to form occipital and adjacent parts of the cerebrum
 - ◆ The hemangiomas represent malformations of the blood vessels
 - ◆ the basic lesion of SWS involves ipsilateral angiomas (vascular tumors) of the skin, meninges, and choroid
- Clinically these patients present in many ways , they are

1. Cutaneous Manifestations

Facial Nevus (Port Wine Stain)

- capillary nevus that is flat and blanches on pressure
- V1 distribution: upper face, superior eyelid, and supraorbital region but may also involve V2 and V3 and cross the midline
- may cause hypertrophy of the involved areas
- may involve the nasopharynx

2. Neurological Manifestations

Seizures

- contralateral partial or secondarily generalized seizures
- usually begin in the first year of life
- increase in frequency and severity
- type, frequency, and severity do NOT correlate with the extent of cutaneous involvement
- recurring Todd's paralysis which requires longer periods to recover with eventual permanent paresis in one third

Hemiparesis

- gradually develops contralateral to the facial nevus
- associated with hemiatrophy of limbs, cortical sensory deficits and hemianopia
- spasticity with pyramidal signs

Mental Retardation

- trend towards progression
- ? role of seizures vs the disease process itself

3. Ocular Manifestations

Fundal Choroidal Angiomas

- affected area is a dark colour
- dilated retinal veins, ectopia lentis, optic atrophy
- homonymous hemianopia (33%)

Others

- iris heterochromia, congenital (buphthalmos) or acquired glaucoma (25-50%)



Fig 1 X-ray of a patient with tram line calcification

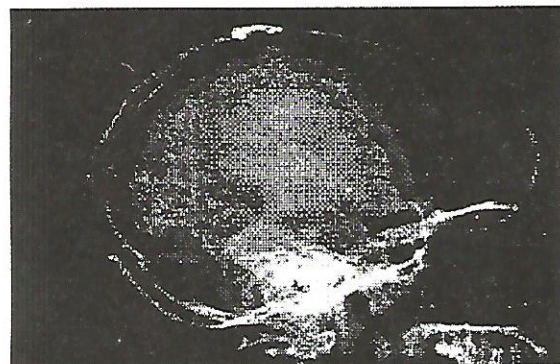


Fig 2 another patient

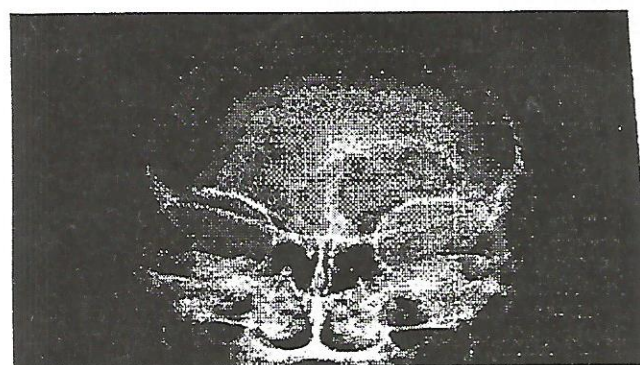


Fig 3 AP Skull x-ray

Conclusion

Plain x-ray skull and CT scan of brain confirm the clinical impression and establish the diagnosis of this syndrome. The plain x-ray findings of the rail road or tram line calcification are very convincing. And after that on non enhanced CT of the brain, typical leptomeningeal venous angiomas confined to pia mater are seen. The Location of

these lesions is more in the parietal then occipital and then frontal lobes.



Fig 4 Another patient with another CT of SWS

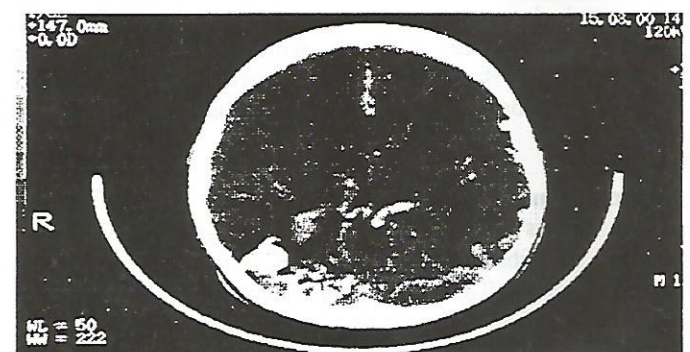


Fig 5 Contrast enhanced CT showing gyral enhancement

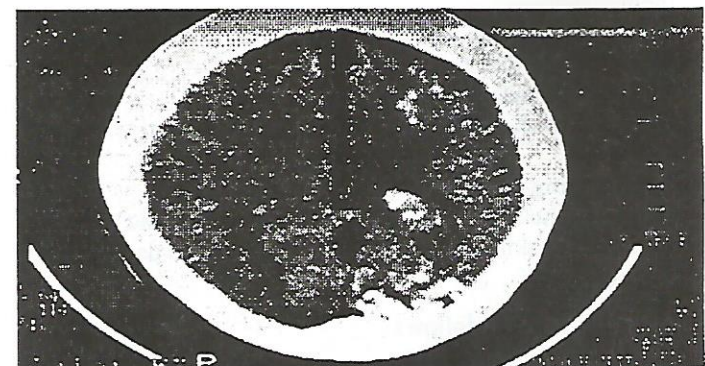


Fig 6 Another occipital Angiomatosis (SWS)

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