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STURGE WEBER SYNDROME: CASE REPORT AND REVIEW

KEYWORDS

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INTRODUCTION:

Sturge-Weber Syndrome (encephelo-trigeminal angiomatosis) is a congenital, rare disorder characterized by a facial birthmark and neurological abnormalities. It is a developmental capillary vascular disorder originating during embryogenesis before neural crest migration, from errors in development of ectoderm and mesoderm in the anterior neural primordium due possibly to somatic mutations.[1]

The most characteristic feature is a "Port Wine Stain" present since birth and involves at least one upper eyelid and the forehead. This varying from light pink to deep purple, is due to an overabundance of capillaries just beneath the surface of the involved skin.

Neurological symptoms appears due to the development of excessive blood vessel on the surface of the brain (angiomas). These are located typically on the occipital region of the brain on the same side as the port wine birthmark. Seizures are the early symptom.

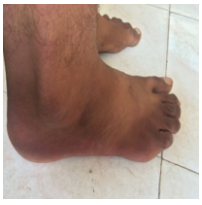
CASE REPORT:

We here by report a case of 24 years old male patient, resident Of Badwani, M.P. admitted in our hospital with complaint of fever associated with chills, headache, vomiting, and burning micturition since 4 days. Patient was a known case of seizure disorder, since childhood and was on maintenance antiepileptic therapy in form of tab. Carbamazepine (100mg) twice daily. Patient was investigated many times before he came to our hospital.

ON EXAMINATION:-

Pt had single, well defined port-wine color macular lesion of approximately 10*8 cm right side of the face, as well as on lateral side of both feet & on the palate which was present since birth. Patient also had swelling of lips.

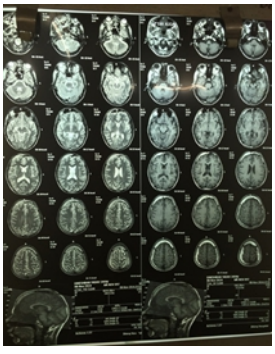
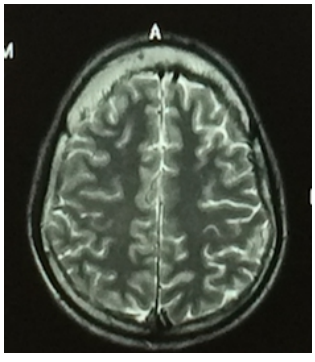
t/p/r – afebrile/78/20
• Bp- 124/76 mmHg
• No pallor/ clubbing/ cyanosis/ icterus/ lymphadenopathy
• Systemic examination R/S- AEBE clear, CVS- S!s2, P/A- soft non-tender no organomegaly, CNS- Higher function, Superficial and Deep Tendon reflexes are within normal limit, planters are flexors bilaterally

- **INVESTIGATION:**

MRI BRAIN	• Hypointense foci on t2w1 are seen right temporal and occipital region- consistant with calcification. • Tortuous dilated vascular channels are noted along left cerebral convexity- p/o pial angiomas. • Focal calvarial thickening noted in left frontal bone. • Mild hypertrophy of right choroid plexus noted.

MRI IMAGES

Discussion

CBC	HB-11.4, TC- 4000, PLATELET- 90000, MCV-112.5, MCH- 39.5, MCHC 35.2
PERIPHERAL SMEAR	Predominantly macrocytic RBCs, normal WBCs with mild lymphocytosis, decreased platelets
MP antigen	Negative
Electrolyte	Na+ - 130, K+ - 4.5, CL- - 95
RENAL FUCTION	Urea- 37, Creat- !.0

Sturge weber syndrome is a congenital nonfamilier syndrome. The Inheritance of Sturge-Weber syndrome is sporadic and it occurs with a frequency of 1:50,000 [2] It manifests as three distinct types,1. Facial capillary malformation(portwine stain) 2. Abnormal blood vessel of brain(leptomeningeal angioma)3. abnormal blood vessels of the eye leading to glaucoma. patient usually presents with seizure, hemiparesis, headache and developmental delay. Cranial CT scan revealing cortical atrophy underlying the angioma with gyriform 'tram-track' calcifications are the characteristic imaging feature. MRI

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is the current gold standard for diagnosis of the disease which is reliable even in very young infants.3There is no specific treatment for this syndrome and it managed symptomatically. Laser therapy can be used to fade portwine stain. The differential diagnosis includes other varieties of arteriovenous malformation as Klippel Trenaunay Weber syndrome, Rendu-Osler Weber syndrome, Bannayan Riley Ruvalcaba syndrome, Divry Van Bogart syndrome and Cobb syndrome.[4]

CONCLUSION:

Diagnosis and treatment of Sturge weber syndrome relies on a detailed history, thorough clinical examination and investigations and which will be helpful to prevent complication.

References

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