Diagnostic Challenge of Sturge Weber Syndrome Phenotype in Camp Setting
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Abstract
Sturge Weber angiomatosis is a non-developmental, rare condition with a vascular hamartomata’s involving the tissues of brain and face. We report herewith a case presenting with recurrent seizures and facial port wine stain in a camp of tribal area in Central India. This patient presentation mimic phenotype of Sturge Weber Syndrome.

Keywords – Tribal health, Port wine stain, Sturge Weber syndrome Phenotype

Introduction
Sturge Weber syndrome comes under group of disorders collectively known as the phakomatoses (“mother spot disease) They consists of congenital hamartomatous malformation that may affect the eye, skin and central nervous system (CNS) at different times. This syndrome is characterized by the combination of of venous angiomas of leptomeninges, face, jaws and oral soft tissue [1]. Sturge Weber Syndrome is the 3rd most common neurocutaneous disease after Neurofibromatosis & Tuberous Sclerosis [2]. Its incidence is 1 per 20,000- 50,000 live birth. Schirmer first described Sturge Weber Syndrome in 1860.Later a detailed description was provided by Sturge in 1879[3]

Leptomeningeal Angioma’s mostly occur unilaterally in parietal or occipital regions. Angioma’s cause an alteration in vascular dynamic which leads to the precipitation of calcium deposition in in cerebral cortex underlying the angioma .Hemiplagia,seizures and mental retardation develop secondary to Angioma the severity of which is governed by the extent of the lesion [4].The cutaneous Angioma’s is known as Port wine stain ,which occurs unilaterally along dermatome of ophthalmic and maxillary division of trigeminal nerve [4].

Case Report - Young boy aged 15 years presented to camp with complaints of repeated attacks of seizures, and pigmentation on one side of face since birth (Fig.1 clinical picture) He is 5 brothers and sisters, him being the youngest of all. Family history and pedigree analysis for 3 generation did not show any such sign/disorder from both on paternal as well as maternal
side. Patient is illiterate from tripbal area of central India. The signs of irreversible mental retardation is evident. There is no thryomegaly. He needs assistance while performing his daily activities. Port wine stain was present on left side of face since birth and is gradually darkening with age. The first seizure was recorded at the age of 6 months and the seizure was gradually worsening with age. No Aggravating or Relieving factors were reported. Patient also has blurring of vision in his left eye. Right eye is apparently normal. The ophthalmoscopic examination is warranted to exclude angiomas in eye and not possible in the camp setting. We suspected the patient to be of Sturge Weber Syndrome phenotype. The online data mining was attempted with OMIM the Sturge-Weber Syndrome; SWS display Phenotype-Gene Relationships. Its chromosome is located on 9q21.2 and may have mosaic, somatic phenotype. The Gene GNAQ locus may display abnormality. The Nonsyndromic port-wine stains (CMC; 163000) are also known to be causes by somatic mosaic mutation in the GNAQ gene.

Further Analysis: This case need further workup by ophthalmological, dermatological and neuroimaging workup to substantiate the further diagnosis.

Figure 01. Illustrating the facial pigmentation since birth.

Discussion

Sturge-Weber syndrome is characterized by an intracranial vascular anomaly in form of leptomeningeal angiomatosis of the occipital and posterior parietal lobes. The most common symptoms and signs are facial cutaneous vascular malformations (port-wine stains), seizures, and glaucoma. These features prevail in our case. The other differential are the Klippel-Trenaunay-Weber syndrome (149000) is sometimes associated with SWS (see Bonse, 1951 and Nonnenmacher, 1955). Debicka and Adamczak (1979) documented Sturge-Weber syndrome in father and son, with son had congenital glaucoma and the father had simple glaucoma. Sujansky and Conradi (1995) surveyed more than 52 patients and develop written questionnaires, telephone interviews, and review of medical records.

The Cognitive impairment in SWS was not related to seizures in previous studies. Hall et al. (2007) reported 6 patients with phakomatosis pigmentovascularis type II, consisting of nevus
flammeus and mongolian spots; 2 patients were diagnosed with Klippel-Trenaunay syndrome, and 3 had features consistent with both Klippel-Trenaunay and Sturge-Weber syndromes.

**Imaging by PET scans** correlate cortical glucose hypometabolism with clinical features in 13 children with unilateral cerebral SWS, Lee et al. (2001)

**Inheritance**

Unlike the other phacomatoses, including tuberous sclerosis (see 191100), neurofibromatosis (see 162200), and von Hippel-Lindau disease (193300), no clear evidence of heredity has been discovered in SWS.

precursors to a larger variety of cell types and tissues, leading to the syndromic phenotype. Five (0.7%) of 669 samples from the 1000 Genomes Project database were positive for R183Q; noting that the reported prevalence of port-wine stains is 0.3% to 0.5%. Shirley et al. (2013) hypothesized that the 0.7% prevalence in that database represented the occurrence of port-wine stains in the population.

Sturge Weber Syndrome is of two types – Complete & Incomplete. Complete when both CNS & facial Angioma’s are present and Incomplete when only one of these is involved and other is spared. Roach scale [5] is used for classification –

**TYPE 1-** Both facial and leptomeningeal Angioma’s ; Glaucoma may also be present

**TYPE 2-** Facial Angioma’s alone; Glaucoma may also be present

**TYPE 3-** Isolated leptomeningeal Angioma’s , usually no Glaucoma .

It is caused by somatic mutations in the GNAQ gene which result in abnormal development of fetal capillary blood vessels and fetal cortical veins do not coalesce resulting in persistent primitive vessels. Mutations occurring at an earlier stage in development may affect a greater variety of precursor cells & lead to Sturge Weber Syndrome. This results in impaired drainage & venous engorgement and as time progresses, venous ischemia causes gliosis, atrophy & calcification.

This syndrome is characterized by –

1. **FACIAL CAPILLARY MALFORMATION (PORT WINE STAIN**
   Distribution of 1st or 2nd division of trigeminal nerve. Flat and pink lesions in newborns.
   The lesion darkens over time to a port wine deep red colour.

2. **LEPTOMENINGEAL CAPILLARY VENOUS MALFORMATION-** also called as leptomeningeal angioma affects the cerebral hemisphere (primarily parietal and occipital lobes) and the eye. Usually unilateral. Often fills subarachnoid space in the sulci, and large venous structures commonly drain into the deep venous system or superficial veins. Underlying parenchyma may be atrophic with multiple calcific granular deposits.

**CLINICAL PRESENTATION-**

1. Seizures- occur in 80% of patients usually begins in early childhood and is often the first sign of sturge weber syndrome. It is typically focal but can be generalised tonic -clonic seizure.
2. Focal neurologic deficit -
•hemiparesis - can occur with seizures. Contralateral to intracranial lesions
3. Intellectual disability occurs in up to 60% patients, lower outcome IQ in pt's with high seizure frequency, early frontal lobe involvement or bilateral lesions.
4. Visual field defects - homonymous hemianopia leptomeningeal capillary venous malformation can affect occipital lobes or optic tracts.
5. Glaucoma - occurs in up to 70% of patients, most common ocular abnormality.

This case was encountered during Health camp conducted by National medicos organization Madhya Pradesh and Chattisgarh in association with DRI at Primary health care setting covering 140 tribal villages within 50km radii of Chitrakoot (Central India) It included a Participation of 90 Doctors and 310 medical students from 9 states. The total patients seen in the camp was around 20,000. We were able to identify one case of Sturge Weber Syndrome out of the total 20,000 patients seen which justifies its rare incidence of 1 per 20,000-50,000 cases. The Treatment modalities include-
1. Anticonvulsants for seizures
2. Surgical treatment for refractory seizures
3. Low dose aspirin to prevent the progression of impaired cerebral blood flow & hypoxic ischemic neuronal injury.
4. Selective photothermolysis for port wine stains
5. Topical medical treatments & surgical treatment for glaucoma.

Prognosis -
Neurologic function may deteriorate with age and prognosis depends on the extent of involvement and age of onset of seizures.

Conclusion: Identification of syndromes in camp setting is possible with existing limitations. The warrants essential referral to tertiary care centre for management and genetic identification of disease in tribal setting.

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