STURGE-WEBER SYNDROME WITH CORPUS CALLOSUM AGENESIS -A CASE REPORT

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ABSTRACT

Sturge weber syndrome is a neurocutaneous disease that manifests with vascular malformations involving the brain, eye and skin. It is a rare disorder that occurs with a sporadic frequency of 1:50000. The condition is characterized by facial nevus, seizures and intracranial calcifications. We present a case of sturge weber syndrome with corpus callosal agenesis; unreported in literature.

An eight year old boy presented with generalized tonic clonic seizures. There was port wine stain over right upper half of the face and eyelid. CT scan of brain showed atrophy of right occipital lobe with a gyriform calcification in parasaggital region. The posterior horns of the lateral ventricles were dilated. The third ventricle was communicating with inter hemispheric fissure suggesting agenesis of corpus callosum.

The angiomatosis might have caused agenesis of corpus callosum either by tissue hypoxia or by mechanical interference of the development.

Key words: Sturge Weber Syndrome, Agenesis Corpus Callosum, Seizures, nevus.

INTRODUCTION

Sturge weber syndrome (SWS) is a rare and sporadic neurocutaneous disease characterized by facial port wine stain (Nevus flammeus), ocular abnormalities (glaucoma & choroidal haemangioma) and leptomeningeal angiomatosis . There is underlying leptomeningeal venous plexus anomaly and lack of cortical venous drainage. This condition is also called encephalotrigeminal angiomatosis as the angiomas involve the leptomeninges and the skin of the face typically in the distribution of ophthalmic branch of trigeminal nerves. The angiomas mostly involve the occipital and the parietal lobes. The child often presents with headache, seizures, mental retardation, developmental delay and various neurological abnormalities. Neuroimaging studies reveal intracranial calcifications, cerebral atrophy and variable degree of developmental anomalies. (Thomas-Shol et al[1]., 2004; Baselga, 2004[2]; Haslam, 2004)[3]. We present here a case of Sturge weber syndrome with agenesis of corpus callosum (ACC), as the association is unreported in literature

CASE-REPORT

An eight year old boy presented with generalized tonic clonic seizures lasting for two hours associated with frothing from mouth and loss of consciousness. He has similar attacks of seizures which started at the age of one year. The child has developmental delay and was mentally subnormal. On physical examination child was semiconscious with a Glassgow coma scale of 9/15. There was a port wine stain involving right upper half of the face and eyelid (Fig 1). There was no lesion on any other part of body. His head circumference was 50 cms. Ophthalmological examination revealed normal fundus without any evidence of raised intra ocular pressure. Rest of the physical and neurological examinations, hematological and biochemical parameters including skull radiographs were within normal limits. CT scan of brain showed (Fig 2) atrophy of right occipital lobe with a gyriform calcification in parasaggital region. The posterior horns of the lateral ventricles were dilated. The third ventricle was communicating with inter hemispheric fissure suggesting agenesis of corpus callosum. The patient was managed conservatively with intravenous fluids and anticonvulsants. Seizure was finally controlled with midazolam infusion. The child was discharged on anticonvulsants therapy and was kept on regular follow up.

DISCUSSION

The presence of facial portwine stain, seizures and intracranial calcifications suggest SWS in the present

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case. However, association with ACC is interesting as it is not reported in literature. Common intracranial anomalies encountered on CT scan are cerebral hemiatrophy, ipsilateral choroid plexus enlargement, abnormal draining veins and altered myelination. (Terdjman et al., 1991)[4] MRI is useful to delineate additional structural and functional anomalies. Perfusion MR imaging and MR spectroscopic imaging can be used to characterize the early hemodynamic and metabolic changes in SWS. MRI also may demonstrate impaired venous drainage in the affected hemisphere, though CT scan may be normal. The venous stasis results in hypo perfusion of the subjacent brain parenchyma, progressive insufficient to meet metabolic demands leading to atrophy, gliosis or demyelination. (Lin et al., 2003))[5] As the corpus callosum is closely related to choroid plexus of the lateral ventricle, the enlargement of the plexus with stasis might have interfered with the development of corpus callosum leading to agenesis. Alternately, the angiomatosis might have mechanically interfered with the development of corpus callosum.

Although exact pathogenesis of SWS is not known. It is thought to be the result of anomalous development of the primordial vascular bed during early stages of cerebral vascularization. Recently it was suggested that hemangioma formation might be associated with somatic mutational events with the loss of heterozygocity of a locus on 5q possibly playing a causative role in this sporadic lesion (Berg et al., 2001)[6]. Another study generates the hypothesis that the lesions are secondary to impaired vascular tone (White et al., 2004)[7]

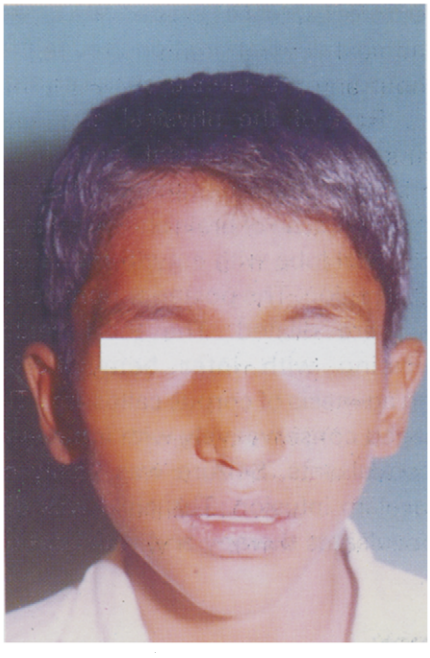

Fig 1. Clinical photograph showing port wine stain along the distribution of right trigeminal nerve

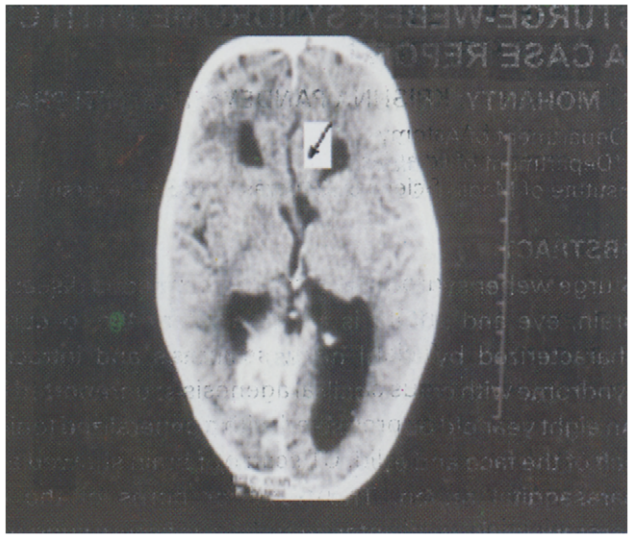

Fig 2. Cranial CT scan showing dense gyriform calcifications in right occipital lobe with dilatation of left posterior horn. Third ventricle is communicating with interhemisheric fissure (arrow).

The development of corpus callosum begins during the fifth week of fetal life with the formation of the primitive lamina terminalis, which thickens to form the commissural plate. Glial cells coalesce to form bridge like structure that serves as guide for callosal fibres crossing the longitudinal cerebral fissure to their targets on the contralateral side of the brain. The mature corpus callosum is formed by the 17th weeks of gestation (Rakic and Yakovlev, 1968))[8].

ACC occurs when the corpus callosum, the band of tissue connecting the two hemispheres of the brain fails to develop normally resulting in disconnected hemispheres. The mechanism of ACC is not clear. It may be an isolated or sporadic finding. Some possible causes include chromosome errors, inherited genetic factors, prenatal infections or injuries, prenatal toxic exposures, structural blockage by cysts or other brain abnormalities and metabolic disorders including inborn errors of metabolism (Jeret at al. 1987[9]; Parrish et al.[10], 1979; Dobyns 1989)[11]. Over 175 genetic syndrome may present partial or total agenesis of corpus callosum (Young 1995))[12].

Associated central nervous system abnormalities includeChiari malformations, anomalies of neuronal migration including lissencephaly, schizencephaly,

pachygyria and polymicrogyria, encepaloceles, Dandy Walker malformations, holoprosencephaly, hydrocephalus, interhemispheric cysts and olivopontocerebellar degenerations(Barkovitch and Norman, 1988)[13]. Extracranial malformations include abnormalities of the face, cardiovascular, genitourinary, gastrointestinal, respiratory and musculoskeletal systems (Parrish et al., 1979;[10] Franco et al., 1993;[14] Kozlowski and Ouvrier, 1993;[15] Lyn 1995). However, association with SWS is unreported in literature. The mechanism of ACC in SWS may be due to hypoperfusion or mechanical interference by the angiomatous tissue during critical period of development.

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