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Schizencephaly – A Rare Case Report

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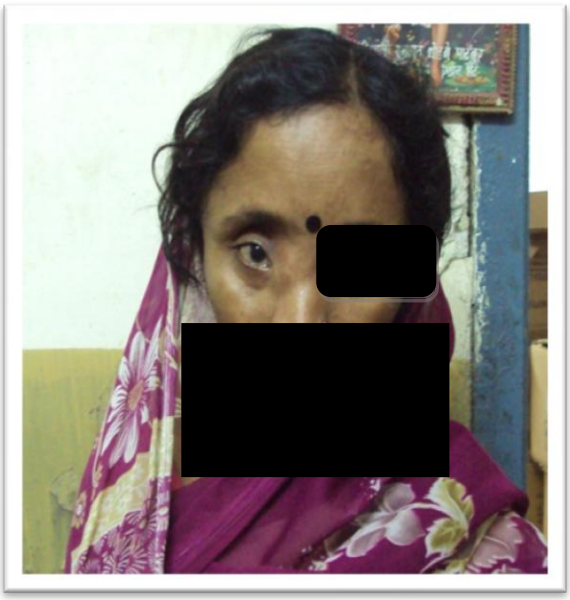
ABSTRACT
Schizencephaly is severe but rare cortical malformation. It is a disorder characterized by a cleft in cerebral mantle, which communicates between the subarachnoid space laterally, and ventricular system medially. Diagnostic modalities are CT scan & MRI. Here we report a case of middle aged female presenting with distinct clinical features & characteristic CT Brain findings. Though the case is rare is a good example for reviewing the literature and coming to diagnosis of schizencephaly.

CASE REPORT

28 Year old Hindu female brought by relatives to the casualty with complaining of generalized tonic clonic convulsions of many years duration with intractable seizure episodes for last 2 days. Patient was in altered sensorium with weakness of left upper limb and lower limb since 2 days.

On enquiry, she was known case of seizure disorder since childhood, the first episode being at the age of 2 months and recurrent such episodes thereafter. She was started on some Ayurvedic medicines details of which were not available with the patients, but she was not investigated. Patient was born of a non-consangeous marriage, full term normal vaginal delivery conducted by Dai at home. The prenatal, intranatal and postnatal period was uneventful.

Baby cried immediately after birth. Mother also gives history of delayed milestones. Walking at the age at of 5 years, Talking at the age of 6 years, Running at the of 7 years, Understanding full sentence at the age of 8 years, Performance in the school was below average. Patient has normal menstrual history and sleep, appetite, bowel and bladder habits were normal.



On Examination patient was conscious, co-operative, comfortable.

Dacrocystitis of right eye is present.No skull, spine, skin or nail abnormality. No neurocutaneous markers were present.

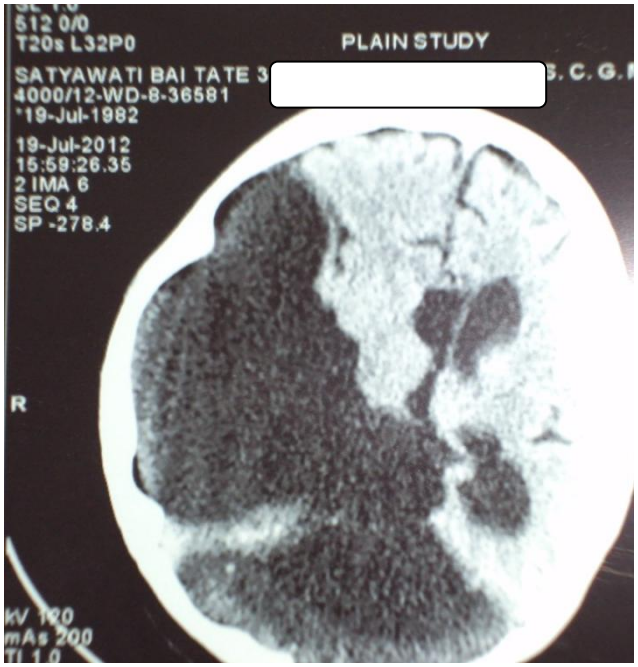
SYESTEMIC EXAMINATION was normal.

On CNS Examination Higher function-normal. Mini mental examination score-26. Cranial nerve examination – WNL. Sensory system examination –WNL. Motor system – Lt hemiparesis with grade 2/5 power in upper limb. 4/5 in left lower limb. Reflexes- WNL expect left planter- extensor. No signs of meningeal irritation

Investigations revealed normal blood reports.

CT BRAIN [PLAIN] :

Large fluid filled cavity occupying right frontal, temporo parietal region lined by grey matter communicated with right ventricle. Finding s/o ***LARGE OPEN CLEFT SCHIZENCEPHALY.***



DISCUSSION

SCHIZENCEPHALY (SPLIT BRAIN)

Listed as RARE DISEASE by office of rare diseases of National institute of health. Incidence is 1.5 /1,00,000 live birth. In literature 70 cases of type 2 schizencephaly has been reported. [3,4] Exact incidence of type 1 is not known.

Yakovlev and Wadsworth coined the term "schizencephaly" in year 1946 [1,2].

Schizencephaly is a rare developmental disorder of the brain characterized by abnormal continuity of histological gray matter tissue extending from ependyma lining of the cerebral ventrical to the pial surface of cerebral hemisphere surface

Types of Schizencephaly

Type I /Fused Cleft /Closed cleft Schizencephaly

It has a cord of gray matter tissue either with no fluid cleft or with ventricular or cortical lips closing one end of an abnormal fluid cleft through the hemisphere.

Type II /Open lip Schizencephaly

It is more common than type I Schizencephaly. It shows a cerebrospinal fluid filled cleft of varying size and shape extending through the hemisphere for the ependyma centrally to the pia peripherally. Associated Neurological malformations are Gray matter heterotropia (collection of gray matter in abnormal location), Polymicrogyria (abnormal brain tissue wit high density of foldings), Arachnoid cysts, Absence of septum pellucidum (80-90%), Cortical dysplasia (seen in contralateral hemisphere in unilateral schizencephaly)

Etiology: The exact etiology is not known. The likely causes may be Genetic (thought to be associated with EMX2 Gene)[5,6.] Physical insult, infection, infarctio, hemorrhage, toxin, mutation

PATHOGENESIS

Schizencephaly is probably a disorder of normal neuronal migration during second trimester of intruterine development, when primitive neuron pre-cursors (germinal-matrix) migrate from just beneath the ventricular ependyma to the peripheral hemispheres where they form the cortical grey matter. Gray matter contains neuronal cell bodies and dendrites whereas white matter contains axons, which are coated in myelin.

PRESENTATIONS

Individuals with clefts in both hemisheres, or bilateral clefts are developmentally delayed, delayed speech and language, corticospinal dysfunction.

Individuals with unilateral clefts presented with hemiparesis, average or near average intelligence, microcephaly, mental retardation, seizures-

generalised tonic-clonic, partial motor, sensory seizures.

Treatment include Physical Therapy, Occupational Therapy, Treatment of seizures, Shunt in case of hydrocephalous .

Complications includes Optic nerve hypoplasia, Skull deformity, Learning disability, seizures.

Prognosis varies depending on the size of neurological deficit. Bilateral clefts are associated with earlier onset of seizures that are more difficult to treat. Patients with open-lip schizencephaly die at an earlier age than those with closed-lip form.

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