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Case Report

Bilateral Schizencephaly with Septo Optic Dysplasia Rare Cause of Seizure Disorder

MMB Zaman

Abstract:

Schizencephaly is an extremely rare developmental birth defect characterized by abnormal slits or clefts in the cerebral hemispheres extending from the lateral ventricle to the cerebral cortex. The margins of the cleft are lined with heterotropic, dysplastic gray matter. The causes of schizencephaly are heterogenous and include teratogens, prenatal infarction/infections, maternal trauma, or EMX2 mutations. It is a central nervous system disorder with variable presentations. People with this disorder commonly have developmental delays, delays in speech and language skills, seizures disorder and problems with brain-spinal cord communication. This condition is present at birth and manifests early in life. This patient presented with seizure and growth retardation and investigation revealed bilateral Schizencephaly with Septo optic dysplasia.

Key words: Schizencephaly, Clefts, Septo Optic Dysplasia.

Introduction:

Schizencephaly is a rare cortical malformation that manifests as a gray matter-lined cleft extending from the ependyma to the pia mater. In 1946, Yakovlev and Wadsworth first described schizencephaly as hemispheric clefts in the region of the primary fissures, infolding of gray matter along the clefts, and associated cerebral malformations. The schizencephaly clefts are mostly perisylvian or centrally located[1].

Schizencephaly is of two types: Type I (closed-lip) schizencephaly is characterized by gray matter-lined lips that are in contact with each other and Type II (open-lip) schizencephaly has separated lips and a cleft of CSF, extending to the underlying ventricle[2].

Exact pathogenesis is not known, but an ischemic episode occurring at the 7th or 8th week of gestation has been hypothesized as an etiological factor. At the 8th week of gestation, neuronal migration starts to form the cerebral cortex from the germinal matrix. These primitive cells begin to migrate along radially oriented glial cells to the cerebral cortical regions. During this period, any insult of vessels in the region of germinal matrix may cause hypoxemia and infarction with arrest of migration of these neuroblasts[3].

1. Dr. M. M. Bodiuzzaman, MBBS, FCPS (Medicine), Junior consultant (Medicine), Faridpur Medical College Hospital.

Address of correspondence :
Dr. M. M. Bodiuzzaman, MBBS, FCPS (Medicine), Junior consultant (Medicine), Faridpur Medical College Hospital.

Tel. +88-01763-771144, E-mail:

Other factors such as infection, metabolic disorders, and genetic defects also play an role in the development of schizencephaly. Granata T et al. have reported heterozygous mutations of the EMX2 gene associated with schizencephaly[3].

Since the exact cause of the disorder is unknown, it's hard to pinpoint risk factors. A few possible risk factors include: having a young mother, having certain genetic mutations, having a sibling, especially an identical twin, with schizencephaly, exposure to certain medications or infections that can disrupt blood flow before birth.

Presenting symptoms are quite variable and related to the amount of brain parenchymal involvement. In general, patients presented with hemiparesis, seizures, and developmental deficits. The severity of manifestations depends on the size and location of the clefts. Patients may present in infancy, childhood, or adults. Patients with closed-lip schizencephaly typically present with hemiparesis and/or motor delay whereas patients with open-lip schizencephaly usually present with hydrocephalus and/or seizures. Patients with closed-lip schizencephaly are more likely to have mild to moderate neurologic deficit than those with open-lip type[4].

Holoprosencephaly, arachnoid cyst, hydranencephaly, and porencephaly are included in the differential

diagnosis of schizencephaly. Porencephaly also extends from the cortical surface to the ventricular surface but is lined by gliotic white matter, not gray matter. Some authors consider schizencephaly as true porencephaly[4].

MRI is the investigation of choice because of its superior differentiation between gray matter and white matter. Identification of gray matter lining the cleft is the pathognomonic finding for schizencephaly. Other associated anomalies are mild hypoplasia of the corpus callosum (most common), absence of septum pellucidum and septo-optic dysplasia[4].

As a rule, therapeutic management of both types of schizencephaly is conservative and predominantly consists of rehabilitation for motor deficits and mental retardation. These patients also need treatment for epilepsy. Surgical treatment is undertaken only in some cases with concomitant hydrocephaly or intracranial hypertension[5].

Physical therapists can help to improve gross motor movements, such as ability to stand and walk. They can also help strengthen of arm and leg muscles.

Occupational therapists can help to improve fine motor movements, such as ability to feed by self and get dressed.

Speech therapists can help to speak or swallow more effectively.

Case Report:

A 12 year old boy presented with complaint of delayed development milestones, like poor school performance, inability to keep pace with same age children, unable to communicate with and understand others, cannot interact with other family member, easy fatigability when he plays and occasional episodes of convulsion which is generalized in nature. His birth history was uneventful, normal vaginal delivery (NVD) was done at home at full term pregnancy without any medical assistance and breastfeeding was adequate. He came from lower socioeconomic family. His parents have no formal education; they are alive with good health and absence of consanguinity. Two other siblings are in good health. There is no history of head trauma. He was treated locally by nonmedical persons and did not consult with any registered medical practitioners before this consultation.

On examination he was found to have features of mental retardation like - having trouble with talking, delayed response of answering questions, difficulty in problem solving and difficulty remembering things. Skull was deformed and there was no persistent opening of any fontanlle. All limb muscles were hypotonic but the child can walk, all limb jerks were preserved, no sensory deficit or any cerebellar signs were seen. Other systemic examination revealed no abnormality. Fundoscopy was normal.

Patient was advised for Random blood sugar (RBS), Alanine aminotransferase (SGPT) and Magnetic Resonance Imaging (MRI) of brain. His biochemical report was within normal limit. MRI report showed frontal and parietal lobe atrophy and bilateral cleft extending to temporal and occipital lobe and the diagnosis was bilateral Schizencephaly with Septo optic dysplasia. Patient was treated with several anticonvulsant drugs but response was not adequate at follow up at 6th month.

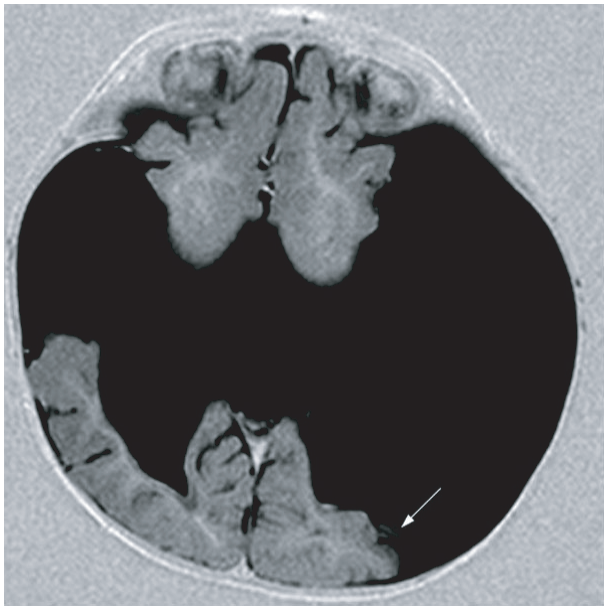

Figure I: MRI images shows Polymicrogyric-dysplastic gray-matter surrounding the clefts bilaterally (arrow).

Discussion:

A case was reported by Shrikant V Rege & Harshad Patil in India[6] of 15-days-old female child. She was referred to neurosurgery department with enlarged head and poor breastfeeding since birth. There was history of seizures and patient was on antiepileptic drugs. Antenatal history was uneventful. Family history was not significant. On examination, macrocephaly with tense fontanelles was present. Scalp was thin and shiny with visible veins. Central nervous system examination revealed spasticity of left upper and lower limbs with brisk deep tendon reflexes. Radiological investigations were performed. Computerized tomography brain showed large fluid density lesion in both frontoparietal

lobes with thinning of brain parenchyma. The lesion appeared to be communicating with lateral ventricle. Magnetic resonance imaging (MRI) brain revealed a large CSF attenuation gray matter-lined cleft present in both fronto- and temporo-parietal region extending from pial surface to ependymal lining of the lateral ventricles. Associated anomalies were partial agenesis of the corpus callosum and absence of septum pellucidum. Based on clinical and radiological features, diagnosis of schizencephaly was made. In my case a boy who presented at about 12 years of age and this is delayed presentation. This is probably due to lower socioeconomic condition, ignorance about medical care of the parents, lack of health awareness, poor health services, and slow development of sign- symptoms.

Another case was reported by P K Chhetri, S Raut, in Nepal[7] in Department of Radiology, a 50 year old male patient was referred for CT scan of the head for a single episode of a generalized tonic clonic seizure. Apart from mental retardation present since birth no other significant history could be elicited. A non - contrast enhanced CT scan of the head was performed. Axial and coronal CT scan showed the wide open - lip schizencephaly with communication of the left lateral ventricle and the subarachnoid space on the left side. There was also absent septum pellucidum and partial thinning of the skull vault on the left side (temporoparietal region).

Apart from above two cases, this case presented so late, probably the sign - symptoms of mental retardation was ignored by the family and patient took attention to physician when he developed convulsion.

All cases have similar presentation like generalized seizure disorders. Two cases have mental retardation and skull deformity. None of them have positive family history.

Conclusion:

Schizencephaly can be considered as a differential diagnosis in children who present with cystic lesions in brain and neurological deficit. An early surgical intervention may help in considerable improvement and limit disability in the child.

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