JOURNAL OF CASE REPORTS 2014;4(2):505-507

Closed Lip Schizencephaly

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Abstract:

Schizencephaly is a rare developmental disorder of neuronal migration characterized by early focal destruction of germinal matrix and surrounding brain before cerebral hemispheres are fully formed at one to five weeks of gestation. This condition is present at birth and presents early in life. Imaging modalities like computed tomography brain can be used to confirm diagnosis. A nineteen year old male presented with generalized tonic clonic seizures. He was diagnosed to be having schizencephaly on computed tomography scan. The patient was started on anti-epileptics and his seizures were controlled. Here we present an adolescent patient with schizencephaly presenting with first onset seizures.

Key words: Schizencephaly, Anticonvulsants, Seizures, Neuroimaging, Brain, Humans.

Introduction

Schizencephaly is derived from Greek word "skhizein" meaning "to split", and "enkephalos", meaning "brain". It is a result of embryonic neuronal migration defect. The cardinal neuro-pathological features include: hemispheric clefts, communication of subarachnoid space with lateral ventricle medially, infolding of grey matter along the cleft and multiple associated intracranial malformations including polymicrigyria, absent septum pellucidum, optic nerve hypoplasia. The presentation and outcome are variable, it could present as hemiparesis, developmental delay, microcephaly, mental retardation but most patients have seizures [1]. There are two types, closed lip schizencephaly (type I) and open lip schizencephaly (type II). Schizencephaly type II occurs more commonly than type I [2]. The overall prevalence of schizencephaly is 1.54 per 100,000 population [3]. There have been very few cases reported especially in the adolescent or adult age group. Here we report a rare case of left sided closed lip schizencephaly, presenting as new onset seizure disorder in an adolescent.

Case Report

A 17-year-old male presented to the outpatient department, with complaints of first onset generalized tonic clonic seizures. A detailed history was taken to rule out any underlying cause of seizures. He was born at term, by vaginal delivery, to a non-consanguineous couple with good

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Received: August 30, 2014 | **Accepted:** December 9, 2014 | **Published Online:** December 25, 2014

Conflict of interest: None declared | **Source of funding:** Nil | **DOI:** http://dx.doi.org/10.17659/01.2014.0128

obstetric history. Growth and development was appropriate according to age. No neurological deficit on examination, with other systems within the normal limits. The patient was investigated further for any metabolic cause of seizures. On blood investigations; complete blood picture, electrolytes, and liver function tests were normal.

Electroencephalography showed focal sharp and slow waves on left side, more in the parietal region. CT of the brain showed a grey matter lined CSF cleft in the left parietal lobe extending from ventricle to plial surface suggestive of closed lip schizencephaly. There was evidence of connection between the lesion and left lateral ventricle [Fig.1]. On the basis of CT findings a diagnosis of left sided parietal closed lip schizencephaly was made.

For control of seizures, he was prescribed oral phenytoin sodium 100 mg thrice a day. The patient was started on oral sodium valproate 500 mg once a day and advised to continue phenytoin sodium before discharge. The patient is regularly being followed up, and there has been no episode of seizure.

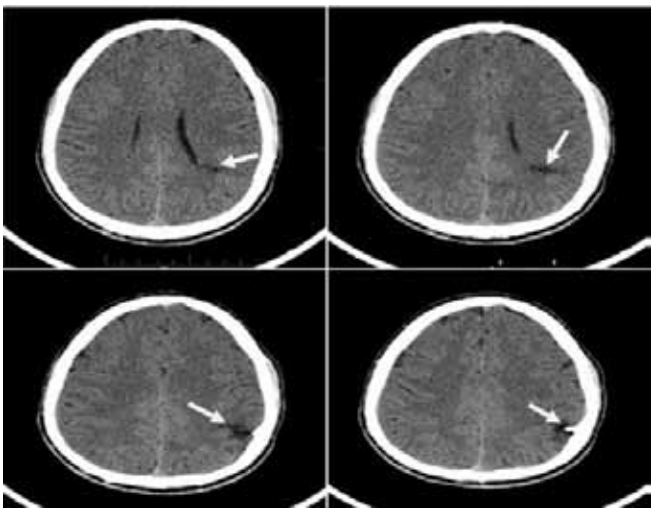

Fig.1: CT scan brain showing cleft in the parietal lobe extending from left lateral ventricle to plial surface suggestive of closed lip scizenchephaly.

Discussion

Schizencephaly is an extremely rare congenital brain anomaly and is the most severe form of neuronal migration defects. This disorder was originally described by Yakovlev and Wadsworth Type I or closed lip type is characterized by cleft walls lined by grey matter with both the lips of the cleft closely apposed to each other with no intervening CSF cavity [4]. Type II or open lip type is a defect characterized by clefts, which are widely separated and communicates with the lateral ventricles [5]. The presence of schizencephalic clefts lined by grey matter suggests that these defects occur early in the second to fifth month gestation, prior to the end of neuronal migration. Etiologies include in-utero infections, cytomegalovirus and herpes virus, maternal trauma of several types, teratogens, alcohol and drug abuse, warfarin, and monozygotic twin interactions [6]. Role of gene Ethyl methane sulfonate (EMX) - 2 mutations is controversial [7]. In our case, there were no stigmata of congenital infections. The antenatal profile was normal. The spine and cranium were normal. Parents of the child were also phenotypically normal.

Clinical presentation depends on the size and location of the lesion. It can have varying effects on neurological development and overall development. Bilateral clefts are generally associated with quadriparesis and severe cognitive impairment. There is very scant literature on schizencephaly in Indian population. Carefully maintained patient records can help us build the database in Indian population. Imaging like CT and MRI are definitive for diagnosis. It should be stressed that schizencephaly can have normal early development and present late in life with non-specific symptoms. It is important to recognize this disease early by prompt imaging like CT or MRI, so that an early diagnosis can be made and multi-disciplinary approach to management instituted.

References

1. Pradhan M, Malla KK, Chowdhury SR *et al*. Case of Schizencephaly: a case report. Kathmandu University Medical Journal. 2003;2:3.
2. Bird CR, Gilles FH. Type 1 schizencephaly: CT and neuropathologic findings. Am J Neuroradio. 1987;8(3):451-454.
3. Curry CJ, Lammer EJ, Nelson V, Shaw GM. Schizencephaly: Heterogeneous etiologies in a population of 4 million California births. Am J Med Genet A. 2005;137:187-189.
4. Yakovlev PI, Wadsworth RC. Schizencephalies: A study of the congenital clefts in the cerebral mantle; clefts with fused lips. J Neuropathol Exp Neurol. 1946;5:116-130.
5. Bapuraj JR, Katariya S, Khandelwal NK. Congenital brain anomalies. *In*: Berry M, Mukhopadhyay S, Chowdhury V, Puri S, Gupta AK, editors. Diagnostic radiology paediatric imaging. 2nd ed. New Delhi: Jaypee Brothers; 2004. pp. 313-329.
6. Eyre J. Neurodevelopmental disorders. *In*: Donaghy M, editor. Brain's diseases of nervous System.12th ed. New York: Oxford University Press; 2011. pp. 207-236.
7. Tietjen I, Bodell A, Apse K, Mendonza AM, Chang BS, Shaw GM, *et al*. Comprehensive EMX2 genotyping of a large schizencephaly case series. Am J Med Genet A. 2007;143A:1313-1316.