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Case Report

Closed Lip Schizencephaly: A Case Report

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Abstract

Neuronal migration defects are rare causes of seizure disorder and developmental problems. Schizencephaly the most severe form is an extremely rare entity. Presentation and outcome of schizencephaly are variable, but it typically presents with hemiparesis, seizures and developmental deficits. Usually the severity of symptoms is related to the amount of the brain affected by the abnormality. Appropriate diagnosis of the disease is necessary to avoid incorrect treatment. Here we reported a rare case of closed lip schizencephaly.

Keywords: Schizencephaly, Hemiparesis, Seizure.

1. Introduction

Schizencephaly is a rare congenital disorder of cell migration with defect in sulcation. It is characterized by cleft in cerebral mantle, which communicates between the subarachnoid spaces laterally and ventricular system medially. This disorder was originally described by Wadsworth and Yakolev[1]. Their original work describes schizencephaly to result from failure of normal migration of primitive normoblasts resulting in cerebral cleft. Two types are recognized, which have prognostic significance. In type I or closed-lip schizencephaly, the cleft walls are in apposition and type II or open lip schizencephaly, in which the walls are separated. Schizencephaly type II occurs more commonly than type I [2]. In either instance the cleft is lined by heterotopic gray matter. In most cases, the gray matter along the cleft is polymicrogyric; in some instances, it is more dysplastic than polymicrogyric[3]. The clefts can be unilateral or bilateral, symmetric or asymmetric and can appear anywhere in the brain, although they usually are perisylvian. In unilateral cases, perusal of the contralateral hemisphere is warranted, as subtle clefts of polymicrogyria are common [3].

2. Case Report

A 15-year-old male child presented to our outpatient department, with complaints of inability to speak since birth and seizures since 5 years of age (generalized tonic and clonic type). He was born at term, by vaginal delivery, to a non consanguineous couple. Antenatal, natal, and postnatal periods were uneventful. There was delay of language milestones since birth. Patient had mental retardation and right sided hemiparesis on examination. Hearing was normal. We investigated the child for the cause of seizures and speech delay. Routine blood investigations, electrolytes, and liver function tests were normal. Electroencephalography showed focal sharp and slow waves on left side more in the tempero-parietal area. CT of brain done in our hospital showed left fronto-parietal large csf density lesion measuring 107 x 49 mm x 75 (AP x TR x SI). The lesion appeared to be lined by grey matter. There was evidence of connection between the lesion and left lateral ventricle. There was mild prominence of third and lateral ventricle. Left sided basal ganglia, thalamus and internal capsule were not demonstrated and left side of midbrain and pons appeared mildly atrophic. On the basis of CT findings, diagnosis of left fronto-parietal closed lip schizencephaly, mild atrophic changes in left side of pons and midbrain and mild hydrocephalus was made.

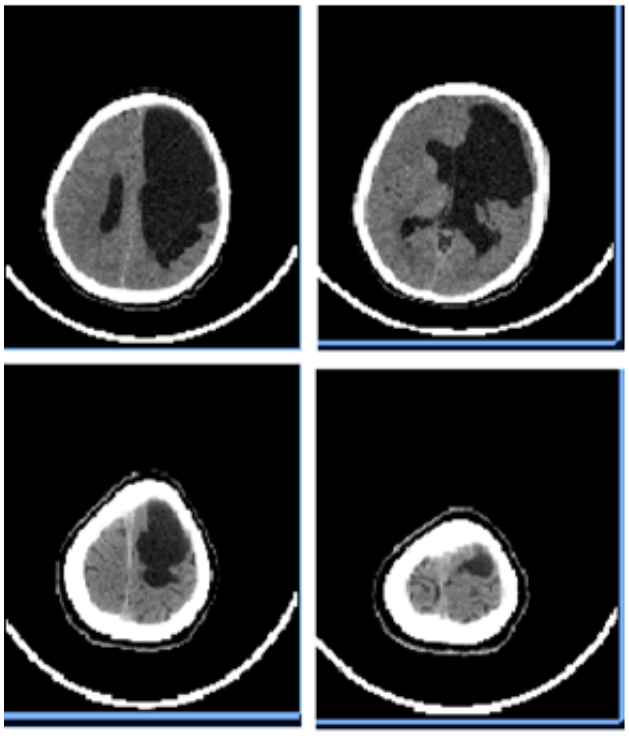

Figure 1: CT Brain shows CSF density lesion measuring 107 x 49 75mm (AP X TR X SI) in left fronto partial region

For seizures, he was not on any medications. The patient was started on oral phenytoin and was discharged. The patient is regularly being followed up, and there has been no episode of seizure till 7 months of treatment of our follow-up. The patient did not make any progress regarding speech, despite speech therapy.

3. Discussion

Schizencephaly is an extremely rare congenital brain anomaly and is the most severe form of neuronal migration defects. Only one-half of the schizencephaly cases are bilateral and when bilateral, only 20% are of mixed type.[4]

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 [5]. Role of gene EMX-2 mutations is controversial [6]. 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 [4]. MRI examination is definitive and is the imaging modality of choice. MRI identifies the anomalous grey matter along the cleft as well as the associated abnormalities such as heterotopias [7]. There is very scant literature on schizencephaly in Indian population. Carefully maintained patient records can help us build the database in Indian population.

References

[1] Denis D, Chateil JF, Brun M, Brissaud O, Lacombe D, Fontan D, Flurin V, Pedespan J. Schizencephaly : Clinical imaging features in 30 infantile cases. *Brain Dev* 2000 Dec; 22(8)475-83.

[2] Bird CR, Gilles FH. Type 1 schizencephaly: CT and neuropathologic findings. Am J of Neuroradio 1987;Vol 8,(3):451-4.

[3] Robertson R, Caruso PA, Truwit CL *et al* Disorders of Brain Development. In: Magnetic Resonance Imaging of the Brain and Spine. Scott W. Atlas. 3 rd Edition. Lippincott Williams & Wilkins, 2002;356-60.

[4] Jones BV. Schizencephaly. In: Barkovich AJ, Moore KR, Grant PE, Jones BV, Vezina G, Koch B, *et al*., editors. Diagnostic imaging pediatric neuroradiology. 1st ed. Utah: Amirsys; 2007. pp. I–36. 39.

[5] Eyre J. Neurodevelopmental disorders. In: Donaghy M, editor. Brain's diseases of nervous System.12th ed. New York: Oxford University Press; 2011. pp. 207–36.

[6] 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:1313–6.

[7] Gusnard DA, Zimmerman RA. Craniocerebral anomalies. In: Lee SH, Rao K, Zimmerman RA, editors. Cranial MRI and CT. 4th ed. USA: McGraw-Hill; 1999. pp. 139–80.