A rare case of schizencephaly in an adult with late presentation

Vijaya Kamble¹, Amol Madanlal Lahoti¹, Avinash Dhok¹, Abhijeet Taori¹, Nilufer Pajnigara²

¹Department of Radiodiagnosis, NKP Salve Institute of Medical Sciences, ²Department of Oral Medicine and Radiology, VSPM Dental College and Research Centre, Nagpur, Maharashtra, India

ABSTRACT

Schizencephaly, i.e., split brain, is a rare, congenital cerebral malformation. Magnetic resonance imaging is the best neuroimaging modality for its diagnosis. In literature, only few cases have been reported causing symptoms in adults. Symptomatic patients present as hemiparesis, developmental deficits to seizures. This condition is usually found at birth and presents during early years. Appropriate diagnosis of the disease is necessary to avoid incorrect treatment. Here, we present a rare case of left, unilateral, open lip schizencephaly in an adult patient presenting with partial seizures but with an asymptomatic childhood.

Keywords: Adults, magnetic resonance imaging, open lip, schizencephaly, seizure

Case Report

A 48-year-old male patient was referred to the Radiodiagnosis Department for MRI brain in view of history of recurrent (three from epilepsy. However, schizencephaly as a cause of seizure in an adult is very rare. No clear-cut gender predilection is noted, but in a study by Stopa et al., they found a slight male preponderance and magnetic resonance imaging (MRI) as an investigation of choice to diagnose schizencephaly and associated anomalies. A study done by Howe et al. found the condition to be more frequent in infants born to younger mothers and is familiar. Various hypotheses have been suggested. It is believed to be because of abnormal neuronal migration or due to localized ischemia. Other causes are the expression of genetic factors such as the mutant gene, EMX2. These expressed genetic factors are believed to damage the periventricular germinal matrix impairing the cellular migration at 6–7 weeks of the intrauterine growth. Sarnat and Curatolo, in their research, described this as an extreme form of true porencephaly due to the ischemic theory. The exact cause of this disorder is still not known.

Address for correspondence: Dr. Amol Madanlal Lahoti, Department of Radiodiagnosis, NKP Salve Institute of Medical Sciences, Lata Mangeshkar Hospital Campus, Digdoh Hill, Hingna, Nagpur – 440 017, Maharashtra, India, E-mail: amollahoti_203@yahoo.co.in

Quick Response Code:

Website: www.jfmpc.com
DOI: 10.4103/jfmpc.jfmpc_43_17

How to cite this article: Kamble V, Lahoti AM, Dhok A, Taori A, Pajnigara N. A rare case of schizencephaly in an adult with late presentation. J Family Med Prim Care 2017;6:450-2.
Kamble, et al.: Schizencephaly in adult patient

episodes) partial seizures for the last 6 months. He had a history of right-sided headache for the past 2 years which relieved after taking oral medications. However, even after medication, the patient complained of lack of activeness and partial weakness on the right side since then. He was symptomatically on the anticonvulsant sodium valproate. There was no history of any episode of seizures in the past apart from seizure episode before 6 months. Family history and birth history were insignificant. The patient then was asked about his birth history which was found normal. His mother had no complications during pregnancy. There were no postnatal complications. There was no global delay in the developmental milestones. Vital signs were normal. In detail, blood and other laboratory investigations were done to rule out metabolic and other causes of seizure.

MRI brain study showed a wide cerebrospinal fluid (CSF)-filled gray matter lined cleft that extends through the cerebral hemisphere on the left side [Figures 1-3]. The cleft extended from the ependyma of left lateral ventricle to the subarachnoid space of left parietal region. Communication of the subarachnoid space with the left lateral ventricle medially with infolding of the gray matter along the cleft was noted.

Right lateral ventricle was normal. Septum pellucidum noted was normal. There was no evidence of hydrocephalus. Corpus callosum appeared normal. Right cerebral hemisphere, cerebellum, pons, and midbrain were found to be normal. A diagnosis of left open lip schizencephaly was made as communication of the left lateral ventricular system with subarachnoid space of left parietal region was noted. The patient was explained about the disease and advised to continue the medical management of sodium valproate.

Discussion

Schizencephaly is gray matter lined cerebrospinal-filled clefts that extend from the ependymal surface to the pial membrane. There are two types reported, Type I or closed lip and Type II or open lip where the walls are separated from each other and filled with CSF. Type II is more common than Type I with 60% of unilateral schizencephaly being open type.

The clefts can be bilateral or unilateral, symmetrical or asymmetrical and can occur anywhere in the brain. It is, however, more common in the parietal and frontal lobes. Our patient has presented with Type II left unilateral schizencephaly, with the walls of the right parietal lobe cleft being widely separated from each other and not lined by polymicrogyric gray matter and having normal intelligence.

Schizencephaly is frequently associated with other congenital abnormalities in 50%–90% of cases such as agenesis of the septum pellucidum and corpus callosum, polymicrogyria-pachgyria (unusually thick convolutions of the cerebral cortex), heterotopias (ectopic gray matter), septo-optic dysplasia, and optic nerve hypoplasia. Our patient was not found with any other associated congenital abnormalities. Presentation and outcome are variable with the severity of symptoms depending on the amount of brain tissue affected. Common presenting features are seizures, hemiparesis mental retardation, delayed milestones, and
motor defects. This patient presented with seizures and weakness of the right side but completely normal developmental milestones and normal childhood. Our case is unusual as very few cases (exact number not mentioned in literature) are reported who have absolutely normal life till (fourth decade) their adulthood and showed symptoms at such later stage of life.

MRI gives excellent demonstration of the anatomic changes and visualization of the gray matter which lines the clefts and it is pathognomonic of schizencephaly differentiating it from porencephaly as well as in identifying other acquired and associated lesions.

This left-sided open lip schizencephaly is the most likely cause of seizure in our patient as other causes of seizure were ruled out clinicopathologically as well as radiologically.

Both types of schizencephaly can be managed conservatively. It predominantly consists of rehabilitation of symptoms such as motor defects and mental retardation and control of seizures. Differential diagnosis of this defect includes focal cortical dysplasia, porencephaly, and band of heterotopic gray matter. Schizencephaly, although a rare congenital disorder, can occur in our environment and can be found even in adult population causing symptoms.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

References
1. Barkovich AJ, Norman D. MR imaging of schizencephaly. AJR Am J Roentgenol 1988;150:1391-6.
2. Yakovlev PI, Wadsworth RC. Schizencephalies; a study of the congenital clefts in the cerebral mantle; clefts with hydrocephalus and lips separated. J Neuropathol Exp Neurol 1946;5:169-206.
3. Denis D, Chatel JF, Brun M, Briassaud O, Lacombe D, Fontan D, et al. Schizencephaly: Clinical and imaging features in 30 infantile cases. Brain Dev 2000;22:475-83.
4. Klingensmith WC 3rd, Cioffi-Ragan DT. Schizencephaly: Diagnosis and progression in utero. Radiology 1986;159:617-8.
5. 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;137:181-9.
6. Stopa J, Kucharska-Miasik I, Dziurzynska-Bialek E, Kostkiewicz A, Zajac-Mnich M, et al. Diagnostic imaging and problems of schizencephaly. Pol J Radiol 2014;79:444-9.
7. Howe DT, Rankin J, Draper ES. Schizencephaly prevalence, prenatal diagnosis and clues to etiology: A register-based study. Ultrasound Obstet Gynecol 2012;39:75-82.
8. Sarnat HB, Curatolo P. Malformations of the Nervous System. 1st ed. Edinburgh: Elsevier Publishers, Science Health Division; 2007. p. 235.
9. Oshiro S, Fukushima T. Two adult cases of unilateral schizencephaly manifesting as minor neurological signs – Importance of radiographic CT assessment. No To Shinkei 2003;55:431-4.
10. Chhetri PK, Raut S. Schizencephaly – A case report. J Coll Med Sci Nepal 2010;6:54-6.