

CASE REPORT

A CASE OF OPEN LIP SCHIZENCEPHALY ASSOCIATED WITH ABSENT SEPTUM PELLUCIDUM AND ARACHNOID CYST

Chris Joseph¹, A. Venkateshwaran², N. Paarthipan³, K. V. Rajasekar⁴

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ABSTRACT: Schizencephaly (spilt brain) is an uncommon disorder of cerebral cortical development, characterized by congenital clefts spanning the cerebral hemispheres from the pial surface to the lateral ventricles and lined by dysplastic cortical gray matter, the condition is present at birth and present early in life. Here we present an adult patient of open lip schizencephaly associated with absent septum pellucidum and arachnoid cyst presenting with seizure on and off for past four years.

KEYWORDS: schizencephaly, septum pellucidum, arachnoid, CT, MRI

INTRODUCTION: Schizencephaly is a brain malformation characterized by in folding of cortical gray matter along a hemispheric cleft near the primary cerebral fissures. This condition was first described by yakovlev and wadsworth in 1946 and the term was given.¹ Currently schizencephaly is described as two types; Type I - closed-lip schizencephaly is characterized by gray matter lined lips that are in contact with each other Type II -Open-lip schizencephaly has separated lips and a cleft of cerebrospinal fluid, extending to the underlying ventricle.²

CASE REPORT: A 26 year old male patient was referred for CT and MRI scan of the head with complaints of headache and giddiness since 6 months duration, associated with few episodes of loss of consciousness and ear blocking sensation lasting for 1 to 2 minutes, throbbing headache which increases with stress along with photophobia. EEG reveals generalized epileptiform activity present.

NCCT and MRI scan of the head was performed. Axial CT scan showed wide open lip schizencephaly (Type I) with communication of right lateral ventricle and subarachnoid space. Associated anomalies were absent septum pellucidum (fig. 4) and an arachnoid cyst. (fig. 6) MRI brain was done and sagittal and Coronal (fig. 1, 2) T2-weighted images showed a cleft in frontoparietal area, lined by abnormal polymicrogyric gray matter, connecting subarachnoid space with lateral ventricular. Associated anomalies like absent septum pellucidum (fig. 3) and an arachnoid cyst (fig. 5) as seen on NCCT was confirmed.

DISCUSSION: Different theories have been described in the etiology of schizencephaly. Barkovich and Norman have hypothesized a vascular etiology. They proposed the abnormality results from an infarction in an area of the germinal matrix during the seventh week of embryogenesis. One hypothesis is based on vascular compromise during early neuroembryogenesis.³

Pathologies like infection, metabolic disorders, ischemia, or genetic defects that cause errors in any of the stages of stem cell differentiation, neuronal migration, or cortical organization form cortical anomalies such as lissencephaly, pachygyria, schizencephaly, heterotopia, polymicrogyria, and unilateral megalencephaly.^{4,5} The etiology is unclear, although a primary malformation secondary to a neuronal migrational anomaly is considered most likely.

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The presenting findings vary with the size and location of the lesion, and include seizures, hydrocephalus, developmental delay, language impairment, and motor dysfunction (hypotonia, hemiparesis, quadriparesis). Patients with closed-lip schizencephaly commonly present with hemiparesis and motor retardation, whereas those with open-lip present with hydrocephalus, seizures. Most of the patients have associated anomalies such as neuronal migration disorders like as dysplasia or heterotopia, the absence of septum pellucidum, corpus callosum dysgenesis, septo-optic dysplasia.⁶

CT scans of closed-lip schizencephaly may show only a slight out pouching, or "nipple," at the ependymal surface of the cleft. The full thickness cleft, or pial ependymal seam, may be difficult to detect on CT scans but is easily discernible on MR studies. Open lip schizencephaly has a larger, more apparent gray matter-lined CSF cleft.⁷ CT may show subependymal or parenchymal calcification in many cases, which suggests that one cause of schizencephaly may be intrauterine infection with cytomegalovirus.⁸ MRI is the imaging modality of choice because of its superior differentiation of gray matter and white matter and its ability to image in more than one plane. Identification of gray matter lining the cleft is the pathognomic finding.⁹

MRI shows the abnormal appearance of the cortical mantle along the cleft and the cortex appearing thicker than normal owing to the presence of polymicrogyria. The contralateral hemisphere may also have developmental abnormalities, such as polymicrogyria and subependymal heterotopias.⁸ Mild hypoplasia of the corpus callosum is commonly seen. The septum pellucidum is absent or nearly completely absent in 70%-90% of affected patients. Those having absence of the septum pellucidum, 30-50 % will have optic nerve hypoplasia on clinical examination. Therefore, septo-optic dysplasia is, by definition, present in 20-45% of patients with schizencephaly.¹⁰ Optic atrophy is usually easily recognizable clinically but is often difficult to confirm on imaging.

Treatment is mostly conservative for seizures, however medically intractable cases may undergo surgical resection. Prognosis depends on size, location, and uni- or bilaterally of the schizencephaly. Patients with closed-lip schizencephaly are more likely to have a mild to moderate outcome than those with open-lip type, and patients with unilateral schizencephaly have a mild or moderate outcome more frequently than those with bilateral lesions.⁶

REFERENCES:

1. Yakovlev P I, Wadsworth R C. Schizencephaly; a study of the congenital clefts in the cerebral mantle. I. Clefts with fused lips. *J Neuropath Exp Neurol*. 5: 116-130, 1946 dic cases.
2. Wolpert, SM and Barnes, PD. *MRI in Pediatric Neuroradiology*. Mosby-Year Book, St. Louis; 1992.
3. Barkovich AJ, Norman D. MR imaging of schizencephaly. *AJR Am J Roentgenol* 1988, 50: 1391-1396.
4. Barkovich AJ, Chuang SH, Norman D. MR of neuronal migration anomalies. *AJR Am J Roentgenol* 1988, 150: 179-187.
5. Kuzniecky RI, Barkovich AJ. Pathogenesis and pathology of focal malformations of cortical development and epilepsy. *J Clin Neurophysiol* 1996, 13: 468-480.
6. S Cakirer, M Beser, K Demir, G M Gallip. Unilateral open lip schizencephaly with absence of septum pellucidum10.1594/EURORAD/CASE.1081

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7. Anne G. Osborn. Disorders of Diverticulation and Cleavage, Sulcation and Cellular Migraton. In: Diagnostic Neuroradiology. Mosby, 1994; 52-4.
8. Gunny RS, Chong WKK. Paediatric Neuroradiology. In: Grainger & Allison`s Diagnostic Radiology. 5th Edition. Volume II. Churchill Livingstone Elsevier, 2008; 1660.
9. Bird CR, Gilles FH. Type 1 schizencephaly: CT and neuropathologic findings. Am J of Neuroradio 1987; 8 (3):451-4.
10. Robertson R, Caruso PA, Truwit CL et al. Disorders of Brain Development. In: Magnetic Resonance Imaging of the Brain and Spine. Scott W. Atlas. 3rd Edition. Lippincott Williams & Wilkins, 2002; 356-60.

Fig. 1 & 2: Sagittal and coronal T2w MRI brain shows the wide open lip schizencephaly with communication of the right lateral ventricle.

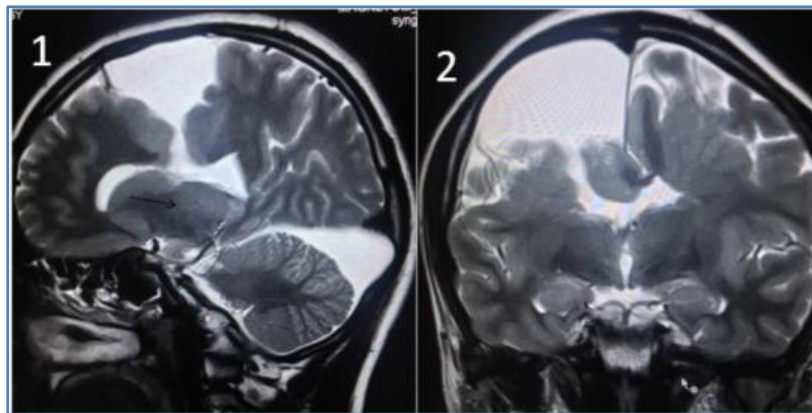


Fig. 1 & 2

Fig. 3: Coronal T2w MRI brain shows the absence of septum pellucidum & Fig. 4: Axial NCCT brain shows the absence of septum pellucidum.

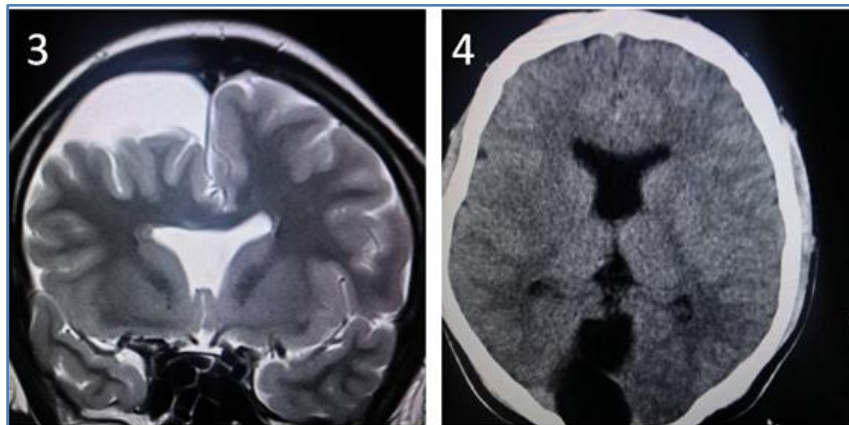


Fig. 3 & 4

Fig. 5: Axial T1weighted MRI brain shows the arachnoid cyst (hyointense) in the posterior fossa and Fig. 6: Axial NCCT brain shows the arachnoid cyst (15HU) in the posterior fossa.

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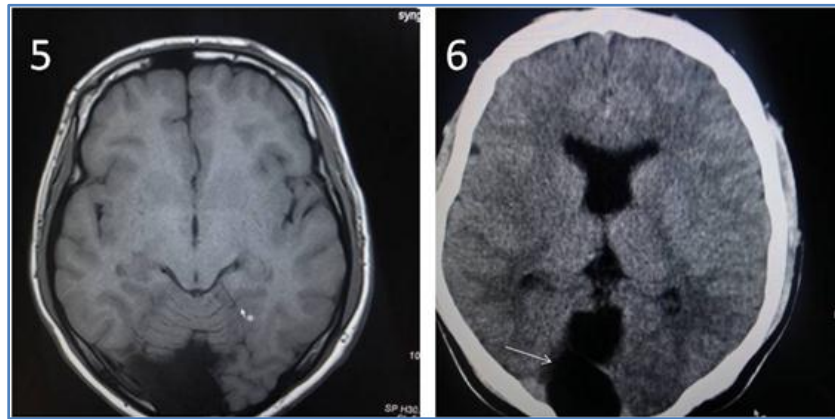


Fig. 5 & 6

AUTHORS:

1. Chris Joseph
2. A. Venkateshwaran
3. N. Paarthipan
4. K. V. Rajasekar

PARTICULARS OF CONTRIBUTORS:

1. Post Graduate, Department of Radiodiagnosis, Meenakshi Medical College and Research Institute, Kancheepuram.
2. Assistant Professor, Department of Radiodiagnosis, Meenakshi Medical College and Research Institute, Kancheepuram.
3. Professor, Department of Radiodiagnosis, Meenakshi Medical College and Research Institute, Kancheepuram.

4. Professor and HOD, Department of Radiodiagnosis, Meenakshi Medical College and Research Institute, Kancheepuram.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. A. Venkateshwaran,
16F, 1-B, Mekarai Appartment,
Annager East Chennai-102
Email: venkateshwaran81@gmail.com

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