Radiology Case Reports 20 (2025) 2690–2693

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Case Report

Unilateral schizencephaly open lip with septo optic dysplasia in adult woman with glaucoma: A rare case ☆

*Eppy Buchori Aristiady, Raisa Mahmudah, Adi Mulyono**

Department of Radiology, Faculty of Medicine, University of Padjadjaran, Dr. Hasan Sadikin General Hospital, Bandung City, West Java, Indonesia

ARTICLE INFO

Article history:
Received 31 December 2024
Revised 10 February 2025
Accepted 16 February 2025

Keywords:
Nilateral schizencephaly open lip
Septo optic dysplasia in adult
Glaucoma

ABSTRACT

This case report presents a 38-year-old female with unilateral open-lip schizencephaly coexisting with septo-optic dysplasia. The patient lacked motor deficits, maintaining normal developmental milestones. schizencephaly is a rare cortical malformation characterized presence of abnormal cleft in the cerebral hemispheres of the brain. These clefts extend from the surface of the brain to the ventricles. Lissencephaly can be unilateral and bilateral and divided into open lips and closed lip. It is frequently associated with other anomaly such as septo-optic dysplasia, grey matter heterotopia, septum pellucidum, and dysgenesis of the corpus callosum. Schizencephaly has no known gender predilection, and estimated incidence of 1.5: 100,000 live birth. Radiological imaging is the cornerstone of diagnosis schizencephaly.

Introduction

Schizencephaly is an rare, congenital disorder affecting cerebral malformation characterized by clefts in the cerebral cortex, which extends from the surface of the ventricle through the white matter. It was first described in 1887, and 2 types exist: closed lip (Type I) and open lip (Type II). Type II is more common, especially in unilateral cases. Magnetic resonance was more sensitive than computed tomography in detection. Schizencephaly is associated in some cases with microcephaly, hydrocephalus, or other malformations such as septo-optic dysplasia and affects the posterior frontal and parietal lobes, with rare occurrences in the temporal or occipital lobe being disorder can be detected in vivo by ultrasonography [1].

Scizencephaly can be associated with other malformations such as septo-optic dysplasia, grey matter heterotopia, septum pellucidum, and dysgenesis of the corpus callosum. Septo optic dysplasia characterized by underdevelopment of the

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

* Corresponding author.
E-mail address: adithedidi@gmail.com (A. Mulyono).

https://doi.org/10.1016/j.radcr.2025.02.065

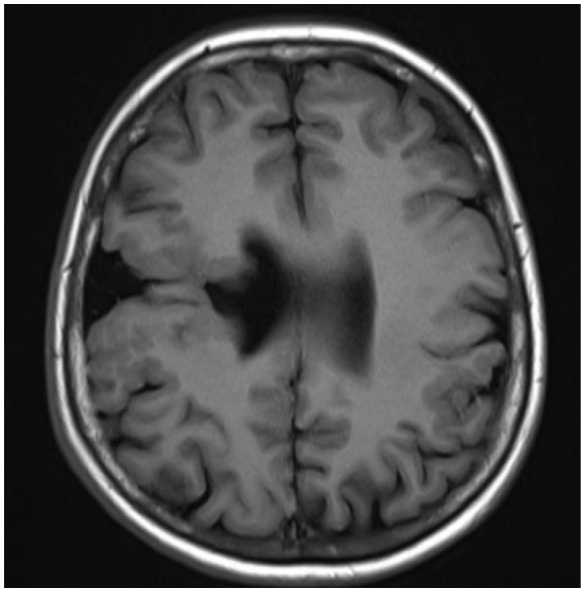

Fig. 1 – Axial MRI T1 imaging showing cerebrospinal fluid signal intensity area in right parietal lobes communicating to the right lateral ventricle.

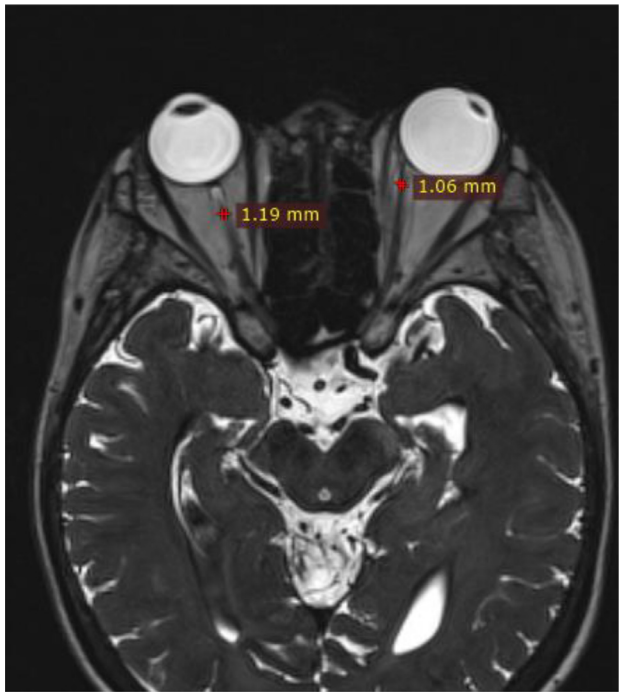

Fig. 2 – Axial MRI T2 imaging showing reduced thickness of the optic nerve sheath.

unilateral or bilateral optic nerve and chiasma, along with the aplasia or hipoplasia of the septum pellucidum, and when associated with schizencephaly, it is referred to as septo-optic dysplasia plus [2].

Case presentation

A 38 year old female patient was referred to the radiology department for an MRI Brain for proptosis since 5 year ago. She had history of blur vision since childhood. She did not have history of seizure. She had no history of limb weakness. There was no family history of neurological or psychiatric disorders, and there was no history of birth-related trauma or maternal exposure to medications. He was a nonsmoker, and nondrinker, and denied any recreational drug use.

On examination, the patient was oriented to time, place, and person, with normal cognitive function. Vital signs were within normal limits. There were no observed abnormalities in growth velocity or pubertal maturation. She had 2 children. Motor examination revealed was normal. Sensory examination was normal. Blood and laboratory test within normal range.

The Brain MRI revealed connection between parietal lobes right lateral ventricle via thin tract with hydrocephalus non communicans. Hypoplasia of optic nerves, absence of the septum pellucidum, thinning of corpus callosum body which features of septo optic dysplasia. The scan also indicated atrophy of right M. rectus medialis also increase size of long axis bulbus oculi bilateral. A diagnosis of unilateral schizencephaly open lip with septo optic dysplasia was made (Figs. 1-5).

Discussion

Schizencephaly is gray matter lined cerebrospinal-filled clefts that extend from the ependymal surface to the pial membrane and occurs mostly in the frontal and parietal lobes in the vicinity of the Sylvian fissure. A study of data from 32 pediatrics revealed that the majority of clefts (42%) were situated frontoparietally [3].

The exact pathogenesis of schizencephaly is not yet clearly understood. There is some hypothesis that explain pathogenesis. In-utero exposure to CMV results in an inflammatory process that leads to a middle cerebral artery (MCA) stroke because the fact that majority of schizencephaly lesion occur in MCA territory. Environmental factors like maternal age (below 20 years), no first-trimester antenatal medical care, and drug or alcohol abuse can contribute to the maldevelopment of the brain in schizencephaly [4]. Development schizencephaly after germinal matrix hemorrhage and PVI in an extremely premature infant born before 24 full weeks of gestation [5].

Schizencephaly can be accompany with agenesis of the septum pellucidum and corpus callosum, polymicrogyria-pachygyria (unusually thick convolutions of the cerebral cortex), heterotopias (ectopic gray matter), septo-optic dysplasia, and optic nerve hypoplasia. In patient with septo optic dysplasia usually associated with small optic disc area and reduce number of nerve fiber, which contribute to blur vision of the patient [6]. Usually, schizencephaly presents in early childhood, typically before the age of 10, during investigations into the causes of seizure disorders or unexplained neurodevelopmental delays. However, instances of schizencephaly emerging in adulthood are rare [7].

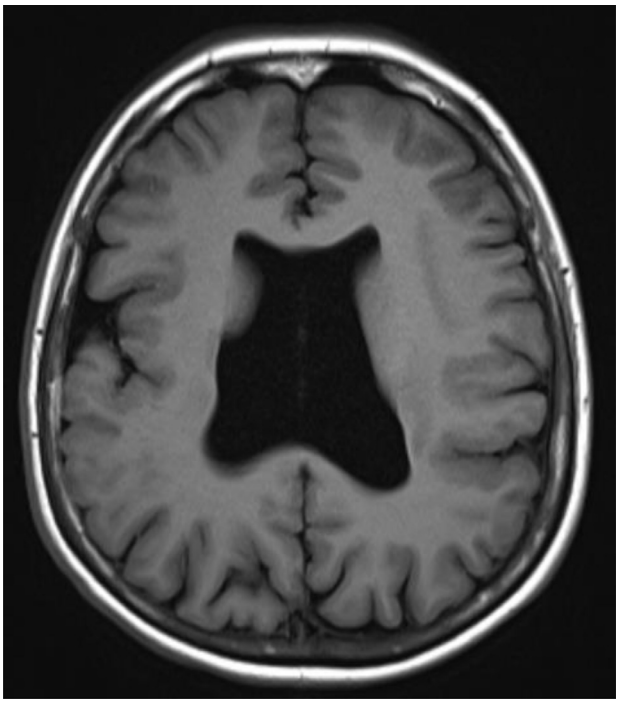

Fig. 3 – Axial T1 weighted MRI image at the level of basal ganglia showing the absence of septum pellucidum.

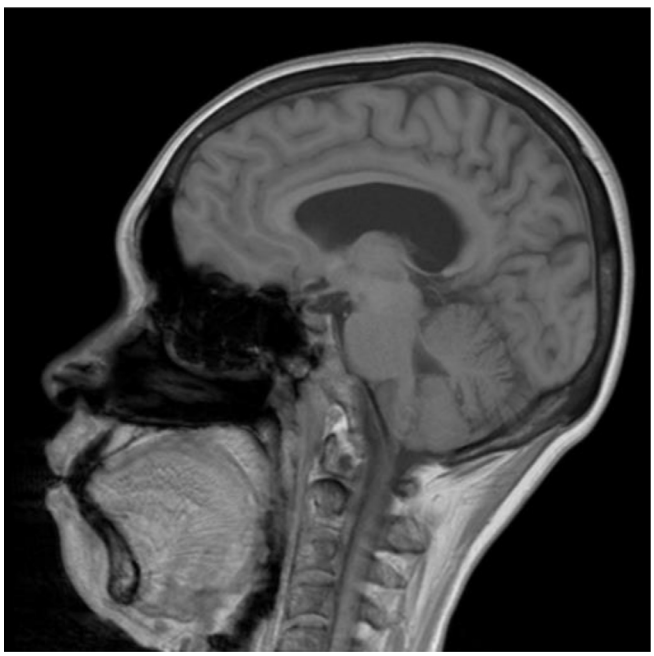

Fig. 4 – Sagittal T1 weighted MRI image showing thinning of the corpus callosum.

MRI is the modality of choice for schizencephaly, showing a fluid-filled cleft with gray matter lining. It distinguishes it from other CNS abnormalities. MRI is also the best modality for the septo-optic dysplasia [8]. n schizencephaly, T2 sequences reveal polymicrogyria, and T1 weighted images and

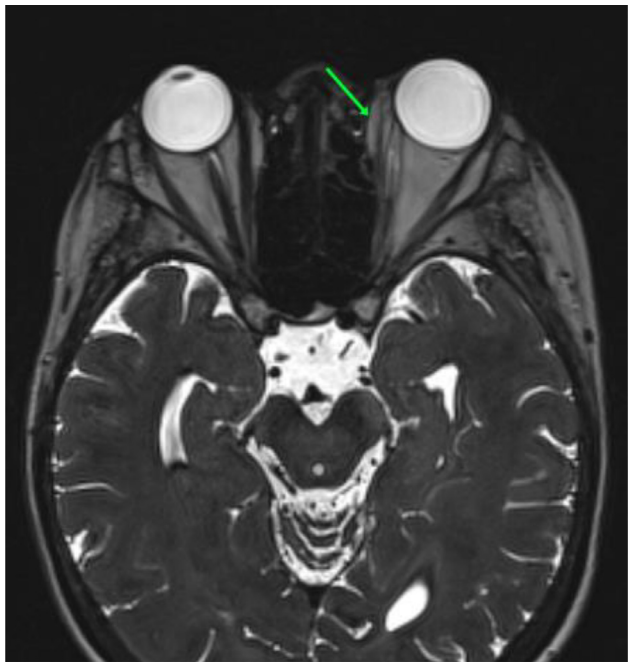

Fig. 5 – Axial MRI T2 imaging showing reduced thickness of the left M. rectus medialis.

FLAIR sequences differentiate white and grey matter. For septo-optic dysplasia, key MRI sequences—T1/T2 axial and coronal for septum pellucidum, pituitary stalk, and ventricles [9].

Both types of schizencephaly can be managed conservatively. 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.

Patient consent

Written informed consent for publication of their case was obtained from our patient.

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