Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Schizencephaly: A Rare Cause of Late-Onset Epilepsy in an Adult

K. Outaghyame[1*], O. Kanali[1], H. Chenter[1], Y. Bouktib[1], A. El Hajjami[1], B. Boutakioute[1], M. Ouali Idrissi[1], N. Cherif Idrissi[1]

[1]Radiology Department, Er-Razi Hospital, Mohamed VI University Hospital, Marrakech, Morocco

DOI: https://doi.org/10.36347/sjmcr.2024.v12i10.013 | **Received:** 21.08.2024 | **Accepted:** 28.09.2024 | **Published:** 02.10.2024

***Corresponding author:** K. Outaghyame
Radiology Department, Er-Razi Hospital, Mohamed VI University Hospital, Marrakech, Morocco

Abstract **Case Report**

Schizencephaly is a rare congenital brain malformation characterized by a cleft that extends through the entire cerebral hemisphere, from the lateral ventricle to the cerebral cortex. This condition can result from both genetic and environmental factors. Although typically identified at birth or during early childhood, adult presentations are rarely documented in the literature. Clinical manifestations vary based on the size and location of the lesion, commonly including hemiparesis, developmental delays, or seizures. Neuroimaging, especially MRI, is crucial for diagnosing schizencephaly and ruling out other potential causes. Accurate diagnosis is essential to avoid inappropriate treatment. We present the case of a 35-year-old patient with partial epileptic seizures who was asymptomatic throughout childhood, found to have unilateral open-lip schizencephaly. This case emphasizes the role of neuroimaging, particularly CT scans, in diagnosing schizencephaly and identifying any associated abnormalities.

Keywords: Schizencephaly, Seizure, CT scan, Adult.

Copyright © 2024 The Author(s): This is an open-access article distributed under the terms of the Creative Commons Attribution **4.0 International License (CC BY-NC 4.0)** which permits unrestricted use, distribution, and reproduction in any medium for non-commercial use provided the original author and source are credited.

INTRODUCTION

Schizencephaly is a rare neuronal migration disorder characterized by congenital clefts that extend from the pial surface to the lateral ventricles, lined by cortical gray matter. This condition was first described in the late 19th century. In 1946, Yakovlev and Waldsworth introduced the term "schizencephaly" to define a malformation that arises from abnormal brain development, rather than the loss of mature brain tissue, as seen in porencephaly, based on their research on cadavers. The clinical manifestations of schizencephaly vary widely and are closely associated with the extent of the cleft. Children with unilateral clefts often present with hemiparesis and mild cognitive delays, while those with bilateral clefts tend to have more severe mental disabilities and tetraparesis. Interestingly, the severity of epilepsy in these patients does not directly correlate with the degree of the malformation. The exact cause of schizencephaly remains unclear. Both genetic and environmental factors have been implicated, including genetic syndromes (such as tuberous sclerosis), exposure to toxins (such as fetal alcohol syndrome), non-infectious vascular disruptions (like early fetal hemorrhage), and infections (such as cytomegalovirus or vasculitis). According to a population-based study in the United States, schizencephaly has a prevalence of 1.54 per 100,000 people. Globally, it affects approximately 1.5 per million live births, with 1 in every 1650 children affected by epilepsy. While it is rare, schizencephaly can occasionally result in epileptic seizures in adulthood. Most cases are sporadic, with no significant gender preference. We present the case of a 35-year-old patient who experienced partial epileptic seizures despite having an asymptomatic childhood, diagnosed with unilateral open-lip schizencephaly. This case emphasizes the critical role of neuroimaging, particularly CT scans, in diagnosing schizencephaly and identifying related anomalies.

OBSERVATION

A healthy 35-year-old woman was referred to the Radiodiagnosis Department for a brain CT scan due to a history of recurrent partial seizures occurring twice over the past two weeks. The patient reported no significant family history of neurological or psychiatric conditions. She was a non-smoker, did not consume alcohol, and denied any history of recreational drug use or seizures during childhood. Inquiry into her birth history revealed that it was normal, with no complications reported during her mother's pregnancy or postnatally. Additionally, there were no delays in her developmental milestones. Upon physical examination,

the patient appeared alert and oriented, showing no signs of distress, and her vital signs were within normal limits. A thorough neurological examination yielded unremarkable findings. Comprehensive blood and laboratory tests were performed to exclude metabolic and other potential causes of her seizures. The CT scan revealed a right parieto-temporal cleft filled with cerebrospinal fluid, which communicated with the ipsilateral lateral ventricle and extended into the open fissure subarachnoid spaces, leading to a displacement of the lateral ventricle on the same side. This finding was accompanied by agenesis of the corpus callosum and septum pellucidum, resulting in dilation of the occipital horns, giving a characteristic "bull's horn" appearance known as colpocephaly.

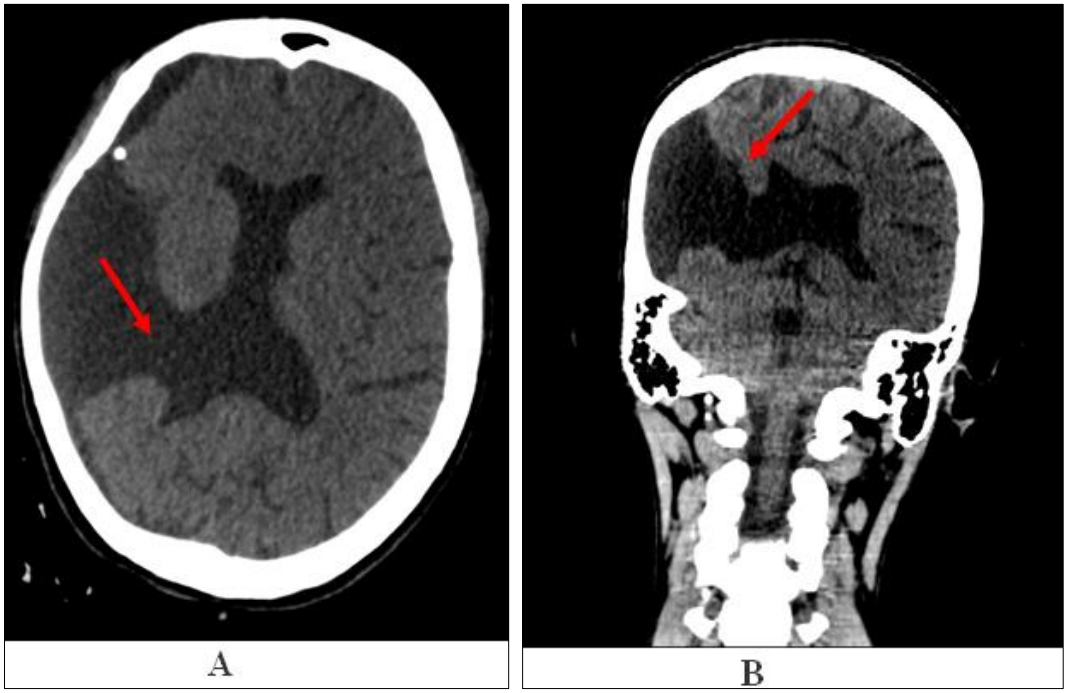

Fig. 1: Cerebral CT scan without contrast injection in axial (A) and coronal (B) sections, showing a right parieto-temporal cleft containing cerebrospinal fluid communicating with the ipsilateral lateral ventricle and extending into open fissure subarachnoid spaces, resulting in attraction of the ipsilateral lateral ventricle

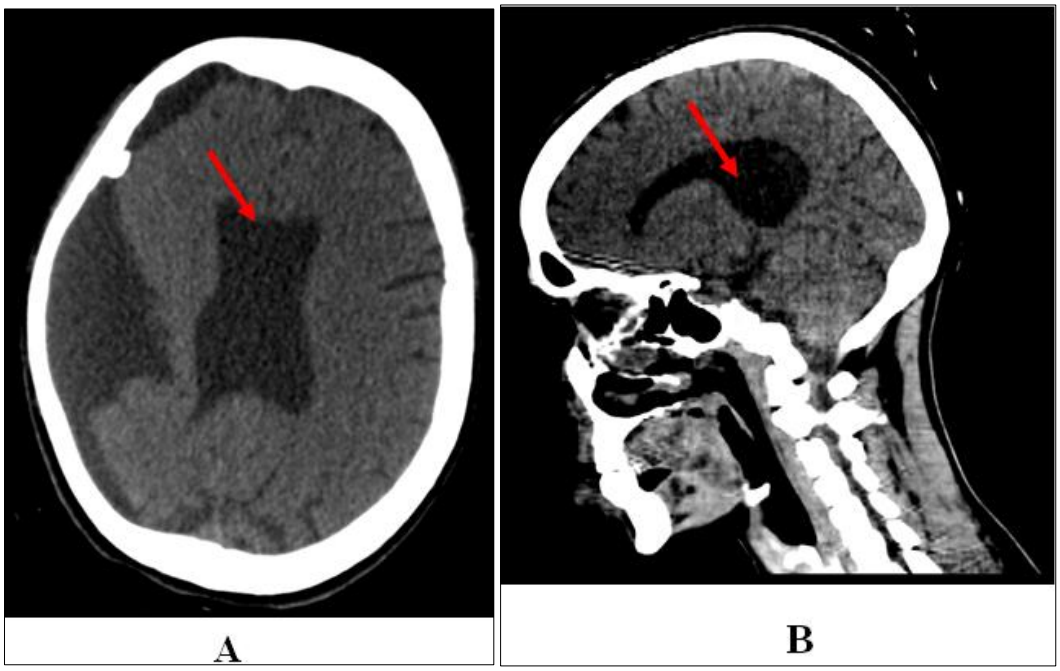

Fig. 2: Cerebral CT scan without contrast injection in axial (A) and sagittal (B) sections, showing agenesis of the corpus callosum and septum pellucidum with dilation of the occipital horns, creating a bull's horn appearance (colpocephaly)

DISCUSSION

The most common clinical manifestations of schizencephaly include epileptic seizures, cognitive impairments, and motor abnormalities. Seizures may be generalized or partial, presenting as simple or complex forms. There are also reports of atypical absences, including West and Lennox-Gastaut syndromes. Both heterotopia and cortical dysplasia are known contributors to seizure activity. The prevalence and severity of cognitive and motor deficits are closely linked to the number of lobes affected, the type and size of the clefts, and the presence and type of associated brain abnormalities. Patients with small unilateral closed-lipped clefts often exhibit milder clinical symptoms, whereas those with large bilateral open-lipped clefts tend to display more severe manifestations. As a result, symptoms can vary significantly, ranging from severe hypotonia and developmental delays in individuals with bilateral clefts to focal seizures that may arise during late infancy or even adulthood in those with unilateral involvement. In cases with large open clefts, abnormal head enlargement due to worsening hydrocephalus may serve as an initial indicator leading to clinical diagnosis. Our patient experienced left-sided seizures while having a completely normal developmental history, making this case particularly unique. It represents one of the very few documented cases—specific numbers are not stated in the literature—of individuals who led a normal life into adulthood (the fourth decade) and began exhibiting symptoms at such a later stage. Recent classifications have identified three types of schizencephaly, with one type not necessarily requiring a full-thickness cerebrospinal fluid (CSF) gap. Type 1 (trans-mantle) is characterized by an abnormal trans-mantle column of gray matter without a cleft containing CSF on magnetic resonance imaging (MRI). Type 2 (closed lip) features gray matter lips that are adjacent but not touching, despite the presence of a CSF-filled cleft. Type 3 (open lip) involves a cleft filled with CSF, with the gray matter lips being separate from each other. Our patient was diagnosed with Type 2 right unilateral schizencephaly. In 50% to 90% of cases, schizencephaly is often accompanied by other congenital abnormalities, including agenesis of the corpus callosum and septum pellucidum, polymicrogyria-pachygyria (abnormally thick convolutions of the cerebral cortex), heterotopias (ectopic gray matter), septo-optic dysplasia, optic nerve hypoplasia, and ventricular enlargement. Our patient presented with schizencephaly along with agenesis of both the corpus callosum and septum pellucidum. Schizencephaly may also coexist with conditions such as subdural hygromas and arachnoid cysts. MRI is the preferred imaging modality for suspected neuronal migration disorders due to its superior ability to differentiate gray matter and provide detailed anatomical information. MRI images often reveal the connection between the clefts and the lateral ventricles, along with the resulting indentation of the ventricular walls. Although commonly located near the Sylvian fissures, clefts can be found unilaterally or bilaterally, symmetrically or asymmetrically, in various regions of the brain. They can also involve the prefrontal lobes and, less frequently, the temporal and occipital lobes. The gray matter lining the cleft is visible on MRI, which aids in distinguishing it from acquired conditions such as porencephalic cysts and others. Treatment for schizencephaly is primarily conservative, focusing on alleviating cognitive impairments and motor dysfunction. Patients typically require management for seizures as well. Surgical intervention is reserved for a limited number of cases involving concurrent hydrocephalus or intracranial hypertension. The extent of the affected cortical areas significantly influences the prognosis

CONCLUSION

Schizencephaly, while a rare congenital disorder, can manifest in our environment and be present in the adult population, leading to various symptoms. This case underscores the importance of MRI in investigating epileptic seizures, as it plays a crucial role in establishing a definitive diagnosis.

BIBLIOGRAPHY

- Denis, D., Chateil, J. F., Brun, M., Brissaud, O., Lacombe, D., Fontan, D., ... & Pedespan, J. M. (2000). Schizencephaly: clinical and imaging features in 30 infantile cases. *Brain and Development*, *22*(8), 475-483.
- Granata, T., Battaglia, G., Incerti, L. D. (1996). Schizencephaly: clinical findings, R Guerrini, F Andermann, R Canapicchi, J Roger, P Pfanner (Eds.), Dysplasias of cerebral cortex and epilepsy, Lippincott-Raven, Philadelphia, 407-415
- Hino-Fukuyo, N., Togashi, N., Takahashi, R., Saito, J., Inui, T., Endo, W., ... & Haginoya, K. (2016). Neuroepidemiology of porencephaly, schizencephaly, and hydranencephaly in Miyagi Prefecture, Japan. *Pediatric Neurology*, *54*, 39-42.
- Packard, A. M., Miller, V. S., & Delgado, M. R. (1997). Schizencephaly: correlations of clinical and radiologic features. *Neurology*, *48*(5), 1427-1434.
- Wilmarth, W. A. (1887). Presentation to the Philadelphia neurological society. *J Nerv Ment Dis*, 395-407.
- Yakovlev, P. I., & Wadsworth, R. C. (1946). A study of the congenital clefts in the cerebral mantle: I. clefts with fused lips. *Journal of Neuropathology & Experimental Neurology*, *5*(2), 116-130.
- Yakovlev, P. I., & Wadsworth, R. C. (1946). Schizencephalies: a study of the congenital clefts in the cerebral mantle: II. Clefts with hydrocephalus and lips separated. *Journal of Neuropathology & Experimental Neurology*, *5*(3), 169-206.