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Case Report

Rare encounter: Adult-onset temporal lobe schizencephaly with septo-optic dysplasia - a case report on comprehensive diagnosis and management☆,☆☆,★

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ABSTRACT

This case report presents a 21-year-old male with recurrent seizures attributed to isolated temporal lobe closed-lip schizencephaly coexisting with septo-optic dysplasia. Despite adult-onset seizures, the patient lacked motor deficits, maintaining normal developmental milestones. Comprehensive diagnostic modalities, including MRI revealing temporal lobe atrophy and associated abnormalities, contributed to the unique identification of schizencephaly. The classic triad of septo-optic dysplasia further complicated the clinical spectrum. Financial limitations influenced the predominantly conservative management, highlighting healthcare challenges. This case enhances our understanding of the rare congenital disorder, emphasizing the importance of tailored diagnostics and management strategies for diverse presentations of schizencephaly, particularly in the context of associated congenital anomalies.

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Introduction

Schizencephaly is an uncommon congenital disorder affecting cerebral cortical development, with an estimated incidence of approximately 1.5 per 100,000 live births. It is characterized by a cleft containing displaced gray matter that connects the cerebral hemisphere to the lateral ventricle [1]. 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. Typically, it affects the posterior frontal and parietal lobes, with isolated occurrences in the temporal or occipital lobe being extremely rare [2].

This condition, occurring between 2 and 5 months of gestation, is mostly sporadic, with disruptions during the seventh and eighth gestational weeks implicated in its etiology [3]. Schizencephaly is associated with other congenital abnormalities, primarily in open-type cases [4]. Septo-optic dysplasia is a neurological condition characterized by the underdevelopment of the optic nerve and chiasma, along with the absence or rudimentary formation of the septum pellucidum, and when associated with schizencephaly, it is referred to as septo-optic dysplasia plus [5].

Clinical presentations vary, ranging from asymptomatic cases to neurocognitive dysfunction, microcephaly, focal seizures, strabismus, spasticity, and seizures. Closed lip schizencephaly may remain clinically quiescent until adulthood [6]. Patients with small unilateral schizencephalies have a good developmental prognosis, particularly when the motor cortex is not involved [7].

Schizencephaly is diagnosed using radiological methods like MRI, CT, or 2-dimensional ultrasonography. MRI shows a fluid-filled linear cleft with heterotrophic gray matter, while CT provides less detailed images [1]. Prenatal suspicion may arise when clefts are visualized within the cerebral hemispheres using two- dimensional ultrasonography [8]. EEG imaging demonstrates the epileptogenic zone, which is often the dysplastic cortex but can be situated not only within the cleft but also in its vicinity and the contralateral hemisphere [6].

Management involves conservative measures for seizure control and rehabilitation. Surgical intervention, such as ventricular shunt insertion, is indicated for concurrent hydrocephalus [1]. In cases of intractable epilepsy, surgical resection of the epileptogenic zone can provide effective seizure control [9].

Case presentation

A 21-year-old male patient was referred to the radiology department for an MRI Brain for recurrent generalized tonic-clonic seizures. He had a history of seizures since childhood and was on antiepileptic medication (sodium valproate). However, his compliance with medication had decreased over the past year, leading to an increase in seizure frequency. In the last 2 months alone, he had experienced 5 seizure episodes. He had no history of limb weakness. He also reported diminished vision in both eyes and mild jerky movements persisting for the past 3 years. Previously, detailed investigations into the cause of seizures had not been carried out. 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 non-smoker, and non-drinker, and denied any recreational drug use.

On examination, the patient was oriented to time, place, and person, with normal development and cognitive function. Vital signs were within normal limits. There were no observed abnormalities in growth velocity or pubertal maturation. Motor examination revealed spasticity in the left lower limb. Sensory examination was normal. The Babinski reflex was negative bilaterally. The ophthalmological examination indicated a corrected visual acuity of 20/40 in both eyes. Pupils measured 4mm in both eyes and responded normally to light. Ocular motility examination detected mild horizontal nystagmus in both eyes.

Further investigations, including blood and laboratory tests, were carried out to rule out metabolic and other causes of seizures, which were normal. The brain MRI revealed right temporal lobe atrophy, a cerebrospinal fluid signal intensity area in the anterior horn of the right anterior temporal pole connected to the right lateral ventricle via a thin tract lined by grey matter (Figs. 1A and B). Mild thinning of the

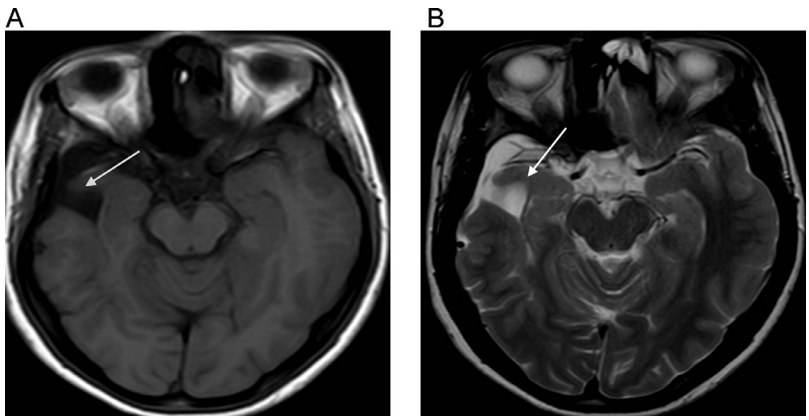

Fig. 1 – Axial MRI image at the level of midbrain showing cerebrospinal fluid signal intensity area in right anterior temporal pole communicating to the temporal horn of right lateral ventricle lined by grey matter (white arrow), as shown in T1 weighted (A) and T2 weighted (B) images.

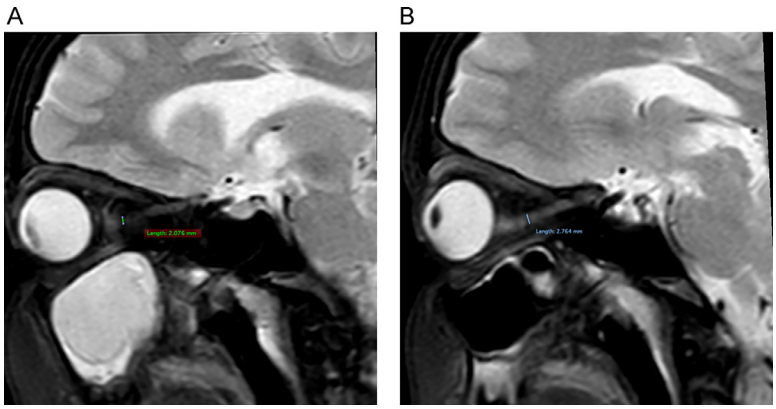

Fig. 2 – T2 weighted oblique coronal MRI images showing the reduced thickness of the right (A) and left (B) optic nerve sheath.

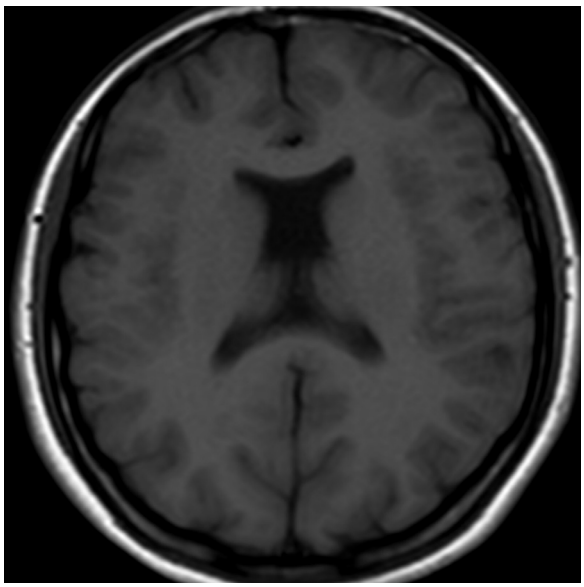

Fig. 3 – Axial T1 weighted MRI image at the level of basal ganglia showing the absence of septum pellucidum.

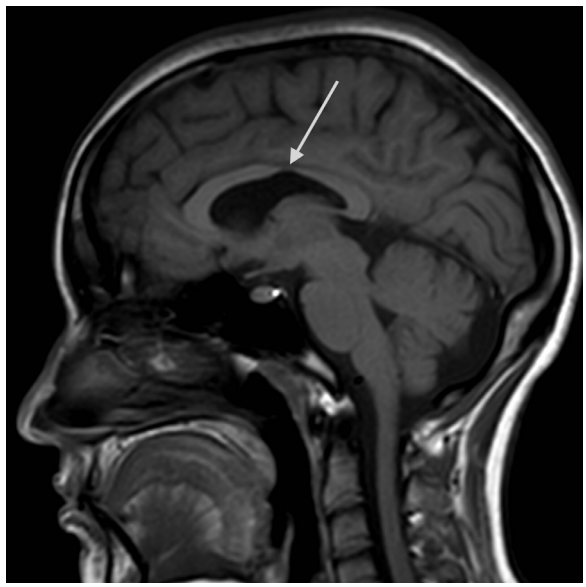

Fig. 4 – Midsagittal T1 weighted MRI image showing the focal thinning of the corpus callosum (white arrow).

retrobulbar region of bilateral optic nerves was observed (Figs. 2A and B). The scan also indicated the absence of the septum pellucidum, focal thinning of the corpus callosum body, and a defect in the anteroinferior portion of the interhemispheric falx, features of septo-optic dysplasia (Figs. 3 and 4). The left cerebral and cerebellum, pons, and midbrain appeared normal. A diagnosis of isolated temporal lobe closed-lip schizencephaly with septo-optic dysplasia was made, and the prognosis was discussed with the patient.

Shunt placement and postoperative physiotherapy were advised. Due to financial limitations, the patient received oral levetiracetam until the surgery. Regular ophthalmological follow-up was also recommended.

Discussion

Schizencephaly occurs mostly in the frontal and parietal lobes in the vicinity of the Sylvian fissure. A retrospective analysis of data from 32 pediatrics revealed that the majority of clefts (42%) were situated frontoparietally [10]. Schizencephaly's etiopathogenesis involves interrupted neuronal migration during gestation, potentially caused by metabolic, infectious, or drug-related factors. Alternatively, vascular compromise in early embryonic stages may induce hypoxemia and infarction, particularly in the middle cerebral artery territory. While isolated temporal lobe schizencephaly is exceedingly uncommon, its occurrence in our case may be attributed to in-utero infection affecting the isolated temporal branch of the middle cerebral artery [8].

Schizencephaly is frequently associated with other congenital abnormalities [4]. Our patient was found to have septo-optic dysplasia. Septo-optic dysplasia (SOD) is a rare congenital disorder characterized by a classic triad of optic nerve hypoplasia, agenesis of septum pellucidum and corpus callosum, and hypoplasia of the hypothalamic–pituitary axis [5].

Schizencephaly is a condition with a variable clinical presentation. A recent comprehensive review by Braga et al. spanning 40 years and encompassing 734 cases of schizencephaly revealed that the predominant and most debilitating issues are motor impairments, affecting around 90% of patients, ranging from hemiparesis to tetraplegia. Neurocognitive impairment is the second most common disability, affecting 77.5% of patients, with more severe cases associated with bilateral clefts. Epilepsy, the third most frequent manifestation, occurs in 67.5% of cases [11]. Usually, schizencephaly presents in infancy or 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. [12]. The patient exhibited seizures without motor deficits and maintained normal developmental milestones throughout childhood. As part of the septo-optic dysplasia spectrum, reduced vision accompanied by horizontal nystagmus was observed.

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 spectrum [7]. In schizencephaly, T2 sequences reveal polymicrogyria, and T1 weighted images and 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; T2 oblique coronal for optic nerve; and sagittal for fornix assessment—are crucial [13].

CT is less detailed for gray matter comparison [7]. In our case, MRI identified isolated closed-lip schizencephaly with communication to the right temporal horn of the lateral ventricle, along with absent septum pellucidum, bilateral optic nerve thinning, and corpus callosum focal defect in the interhemispheric falx.

Given the predominantly conservative approach to management, focusing on seizure control and rehabilitation for motor deficits and mental retardation [1], our patient was administered anti-epileptic drugs to effectively manage seizures. Treatment for Schizencephaly with septo-optic dysplasia (SOD) may encompass hormone replacement therapy for an underdeveloped pituitary gland [14]; nevertheless, given the absence of abnormal growth velocity and pubertal maturation in our case, hormone replacement therapy was not initiated. Additionally, rehabilitation was deemed unnecessary as there were no motor deficits in extremities or cognitive dysfunction detected.

Conclusion

This case highlights the exceptional occurrence of adult-onset seizures associated with isolated temporal lobe closed-lip schizencephaly coexisting with septo-optic dysplasia. The diagnostic precision of MRI and the tailored management approach, highlight the challenges in addressing this rare congenital disorder.

Author contributions

Sundar Suwal: Conceptualization, as mentor and reviewer for this case report and for data interpretation. Shailendra Katwal: Contributed in performing literature review and editing. Dinesh Chataut: Contributed in performing literature review and editing. Suman Lamichhane: Contributed in writing the paper and reviewer for this case. Aayush Adhikari: Contributed in writing the paper. Pratik Baral: Contributed in writing the paper. Amrit Bhusal: Contributed in writing the paper.. All authors have read and approved the manuscript.

Registration of research studies

Not applicable.

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Provinence and peer review

Not commissioned, externally peer review.

Patient consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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