

A Case of Close Lip Schizencephaly with absent septum pellucidum in adult presenting with seizure disorder

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Abstract

Closed cleft lip with absence of the pellucid septum is a rare congenital malformation of the brain that can be associated with a variety of neurological conditions. We report the case of a 25-year-old male with left hemiparesis who presented with recurrent seizures from childhood and increased tremor. MRI brain revealed closed lip schizencephaly with absent septum pellucidum. He had been taking anticonvulsant for last 7 years and is under symptomatic management.

Key Words

Case report | Neurosurgery | Schizencephaly | Seizure Disorder | Septum Pellucidum

Introduction

Schizencephaly is a rare congenital brain malformation characterized by abnormal clefts in the cerebral cortex. Schizencephaly is rare and thought to affect 0.54 to 1.54 out of every 100,000 live births.¹ 1.48 per 100,000 live births is the estimated prevalence.² Patients may present with seizures, developmental delay, and motor deficits. Absent septum pellucidum is a common association seen in patients with schizencephaly and has been reported to be present in up to 50% of cases.³ Diagnosis is typically made through neuroimaging studies such as MRI. We report a 25-year-old adult who has an incidental diagnosis of schizencephaly with absent septum pellucidum.

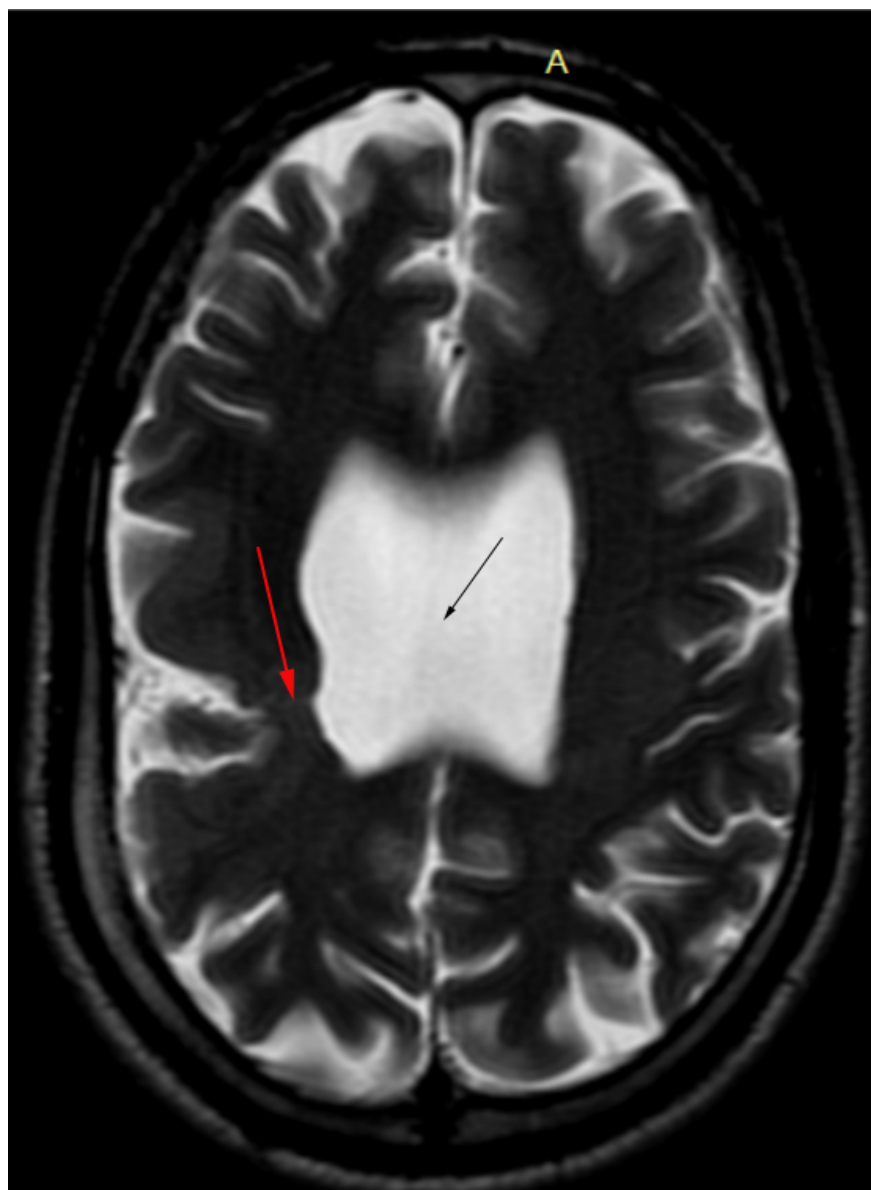
Case Report

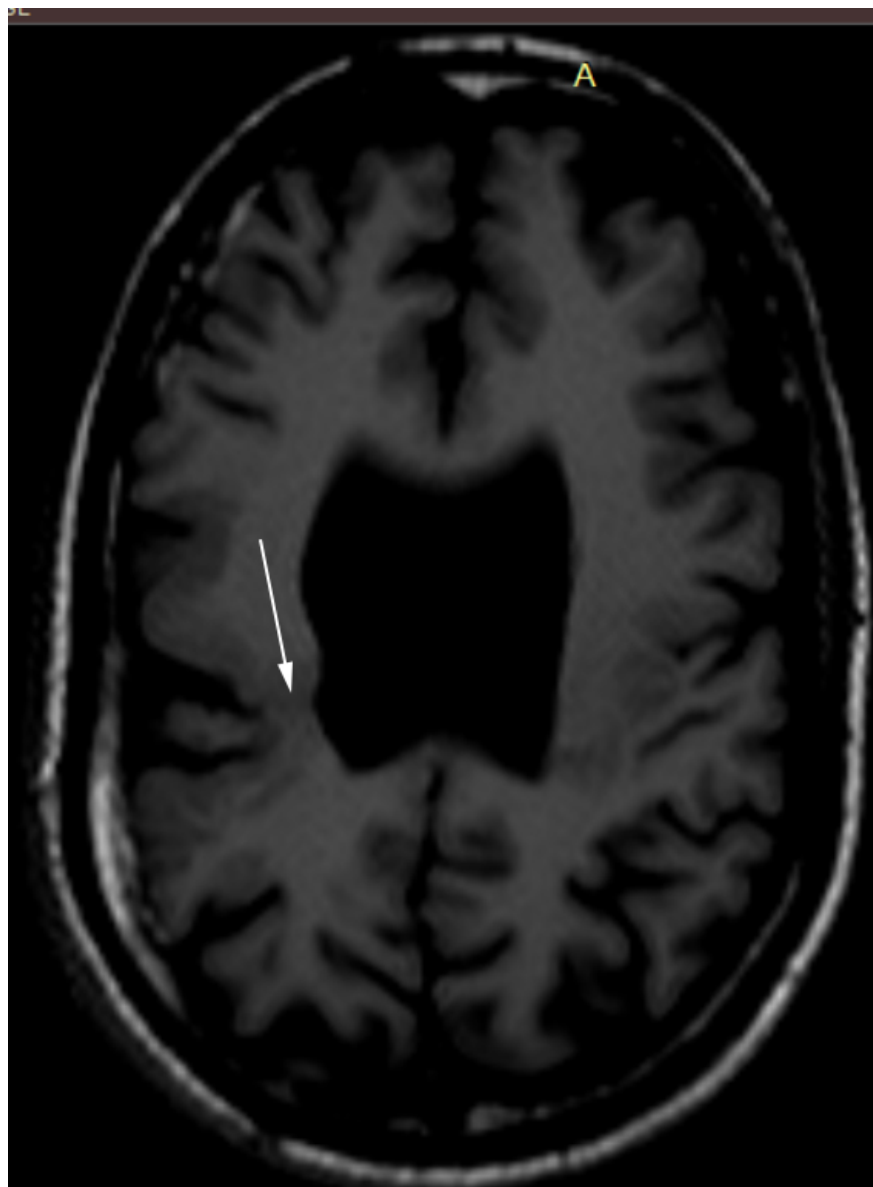
A 25-year-old male presented to the outpatient clinic with a chief complaint of increased tremor in both upper extremities (Right > Left) without any weakness. He had a history of recurrent seizures since childhood for which he was under antiepileptic medication (Sodium Valproate) but workup on cause of seizure was not done in the past. Left sided weakness was present at birth which resolved as stated by the patient. There

was no significant family history of neurological or psychiatric disorders. The patient was unemployed, a non-smoker, non-drinker, and denied any history of recreational drug use.

On physical examination, the patient was alert and oriented, with no signs of distress. His vital signs were within normal limits. Neurological examination revealed high amplitude moderate frequency resting tremor of both upper extremities with left sided weakness (Power 4/5 in both left upper and lower limb). Babinski reflex was negative bilaterally. Considering the possibility of tremor due to Valproate, antiepileptic regimen was changed to Levetiracetam.

MRI brain showed subtle T2 hyper intense curvilinear cleft communicating the lateral ventricle and sub-arachnoid space of right fronto-parietal convexity which is lined by closely apposed grey matter. Absence of the septum pellucidum is noted resulting direct communication between the lateral ventricles and squaring off of the frontal horns. (Figure 1 and 2). MR axial images through the level of optic nerve shows their normal morphology and normal morphology of bilateral globes (Figure 3)







The patient was started on anti-seizure medication for seizure disorder and referred to a neurologist for further evaluation and management of his tremor. Physical therapy was also initiated to address his left hemiparesis and improve motor function.

Discussion

Schizencephaly is a rare congenital neuronal migration disorder characterized by a cleft lined by heterotopic gray matter, which connects the surface of the cerebral hemisphere to the lateral ventricle.⁴ The term was coined by Yakovlev and Wadsworth in 1946, based on their work on cadavers, that classified schizencephaly into two types.⁵ These are:

Type I (closed-lip): Cleft is fused, preventing CSF passage.

Type II (open-lip): A cleft is present, which permits CSF to pass between the ventricular cavity and sub-arachnoid space.

Developmental delays and a variety of neurological impairments can be linked to both kinds of schizencephaly. The etiopathogenesis of schizencephaly remains unclear, but it may result from external factors like middle cerebral artery stroke or genetic factors like EMX2 gene mutation. Maternal age, substance abuse, and lack of prenatal care are also potential risk factors.⁶ A related condition is absent cavum septum pellucidum (CSP), which is characterized by the absence or underdevelopment of the cavity between the two lateral ventricles in the brain. In instances of closed lip schizencephaly, the absence of CSP is a frequent finding,

and it is frequently employed as a diagnostic standard for the syndrome. With the help of several imaging methods, such as magnetic resonance imaging (MRI), the absence of the CSP may be shown.⁷ The cleft in the cerebral hemisphere can be seen through imaging tests like an MRI, which are commonly used to diagnose schizencephaly. Managing the neurological abnormalities and developmental delays associated with the illness is often treated with physical therapy, occupational therapy, and speech therapy.⁸

Also, recent research have looked at the use of fiber tractography and diffusion tensor imaging (DTI) in the diagnosis and treatment of brain abnormalities such schizencephaly.⁹ Although fiber tractography may be used to see the route of important white matter pathways in the brain, DTI is a type of MRI that can give information on the microstructure of brain tissue and the integrity of white matter tracts. These methods could offer insightful details regarding the underlying neurological abnormalities and potential remedies for schizencephalic patients.

DTI and fiber tractography may provide valuable information for the diagnosis and management of these conditions, while genetic testing and molecular profiling may help to identify underlying genetic causes and potential treatment targets.

Some genetic mutations have been reported as possible etiological factors for schizencephaly. The main genes identified in this regard are COL4A1 mutations, EMX2-germline mutations, SHH gene, SIX3 gene.⁴

As the precise source of the disorder is unclear, controlling its symptoms and deficiencies serves as the mainstay of treatment.

Conclusion

To sum up, rare congenital brain anomalies such closed lip schizencephaly and the missing cavum septum can cause a range of neurological deficits and developmental delays. Like in our instance, such deficits may come in light during maturity as a variety of neurological symptoms. The cause of recurrent neurological symptoms should be identified in order to determine the best treatment option and prognosis. This should be done whenever feasible in an environment with plenty of resources.

Author Contribution

Chhabi Khadka : Conceptualization; data curation; methodology; writing – original draft. **Umang Gupta** : Conceptualization; supervision; validation; writing – original draft; writing – review and editing. **Prakriti Bhandari** : Conceptualization; data curation; validation; writing – original draft; writing – review and editing. **Prabin Pandey** : Data curation; methodology; validation; writing – original draft; writing – review and editing. **Shailes Paudel** : Conceptualization; formal analysis; supervision; validation; writing – original draft; writing – review and editing

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Conflict of Interest statement

None declared.

Ethics Statement

Ethical approval was not required for the case report as per the country's guideline.

Informed Consent

Written informed consent was obtained from the patient to publish the report.

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