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CASE REPORT

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# Schizencephaly diagnosed after an episode of seizure during labor: A case report

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**Abstract**

Schizencephaly, an extremely rare anomaly of the cortex, is characterized by abnormal clefts in the cerebral cortex. Very often, this condition is diagnosed early in the childhood period but few instances exist in literature where schizencephaly-associated seizures and hemiparesis have presented later in life too. Here, we report a rare case scenario of a lady in her late 30s who initially presented to us with obstetric concerns wherein schizencephaly remained an incidental finding despite the significantly large cortical cleft along with lobar holoprosencephaly and lissencephaly.

**KEYWORDS**

holoprosencephaly, lissencephaly, schizencephaly, seizure

## 1 | INTRODUCTION

Schizencephaly is an uncommon anomaly of the brain characterized by abnormal gray matter lined clefts extending from the ependymal surface of the ventricle to the subarachnoid space. There are two distinct subtypes. Cleft with fused lips is a type 1 or closed type whereas unopposed lips allowing communication of the cleft to the ipsilateral ventricle is an open type.[1] Although the exact cause of this disorder is uncertain, diverse theories have been put forward to establish a definite underlying etiology. A defect in neuronal migration[2] or some form of molecular genetic abnormalities, such as mutation in EMX2,[1] SHH, and SIX 3 genes,[3] intrauterine infections[4] are few of the probable causes advocated in literature. MRI is the best suited imaging modality for diagnostic evaluations owing to its superiority in gray and white matter distinction, and hence distinguishing the entity from porencephaly or any other post infectious, postoperative, or post necrotic lesions.[2,5]

## 2 | CASE PRESENTATION

A 37-year-old gravid lady presented to the gynecological outpatient department (OPD) of Dhulikhel Hospital at her 38th week of gestation. She complained of lower abdominal pain and vaginal discharge starting from 1 day prior to presentation. She was clinically stable. The fetal heart rate was 132 beats per minute. On abdominal examination, the uterus was of term size with longitudinal lie and presentation was cephalic. Membranes were absent and liquor was clear.

This was her fourth pregnancy. Her previous pregnancies were reportedly uneventful. She had not attended any antenatal care checkups during this pregnancy. The

patient party denied history of any concomitant medical issues as well. There were some inconveniences communicating with the patient, she was uncooperative and did not communicate properly with any health professional, the relevant history was extracted from her husband who had accompanied her to the hospital.

She was admitted to the labor ward for prelabor rupture of membrane. Augmentation of labor was being done with injection syntocin. Her vital signs and fetal heart rate were continually monitored. She was clinically stable but 6 h after admission, developed generalized tonic clonic seizure involving bilateral upper and lower extremities. The fetal heart rate promptly dropped to 80 beats/min. She was immediately attended by a senior consultant. Injection syntocin was stopped. A loading dose of magnesium sulfate injection was given under supervision. Oxygen at 4 L/min was started. An emergency caesarean section was planned. The outcome was a single, alive, male baby of weight 2730 g, with APGAR score 7/10 and 8/10 at 1 and 5 min, respectively. Post operatively, the patient was shifted to intensive care unit where she was assessed by a team of neurosurgeon and internist.

As there were difficulties communicating with the patient herself, she was reluctant to give a detailed history of her illness. Her husband was re-inquired and asked to elicit an elaborated history of her condition. The patient had been having recurrent seizure episodes, but they never sought medical attention for it. And apart from the fits, she had no other issues. We noticed that the patient was not willing to talk to any hospital attendants but was normally conversing with her visitors. Only a limited history could be elicited.

At present time, she has one or two seizure episodes in a month. With some difficulties, a detailed neurological examination was done. She was conscious and well oriented to time, place, and person. Cranial nerves examinations were normal. Reflex was 2+ on both sides. Motor tone was normal, and motor power was 5/5. B/L plantar reflex was downgoing. Sensory examination was normal.

Complete blood count, CRP, ESR, liver function test, renal function test, lipid profile, random blood sugar, serum electrolytes, serum calcium and magnesium, urine analysis were performed. Investigation reports revealed no abnormalities. Routine EEG was unremarkable; however, 24-h EEG was not done. MRI of the brain revealed hypointense cortical cleft in T1 image (Figure 1**)** and hyperintense in T2 (Figures 2 and 4A), suggestive of left-sided open lip schizencephaly. The absence of septum pellucidum with fusion of frontal horn of lateral ventricles (Figure 3) and a hyperintense lesion in the left temporal lobe was also noted (Figure 4B). A screening echocardiographic examination of the baby revealed no abnormalities. Levetiracetam was started and we tried to assess all possible barriers of future drug adherence and made an effort to mitigate those.

## 3 | DISCUSSION

Schizencephaly, a cerebral structure abnormality, occurs mostly in the frontal and parietal lobes in the vicinity of sylvian fissure.[5,6]

Various postulates have been set forth to explain etiology. Detrimental elements causing developmental disorders in the seventh and eighth week of gestation could be responsible. At the 7th week of gestation, neuroblast cells are formed from the germinal matrix, and at eighth week, driven by the radially oriented glial cells, they migrate to form the cerebral cortex.[7] Any form of prenatal insult, be it metabolic, infectious, or drug related during this period can lead to developmental arrest thereby interrupting normal stem cell differentiation

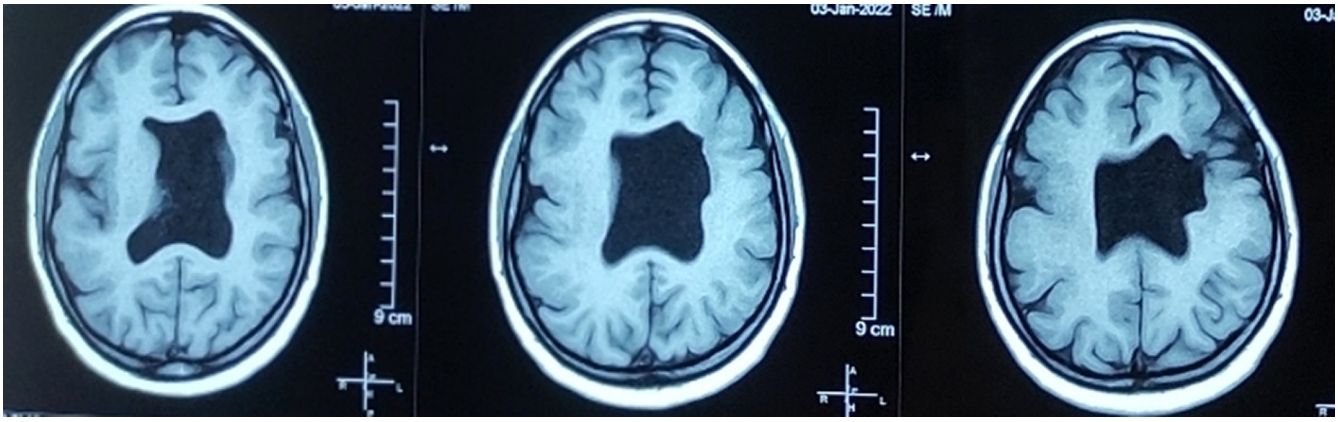


**FIGURE 1** T1 axial view showing gray matter lined hypointense cleft extending from cortical surface of left temporal lobe to body of left lateral ventricle-open lip schizencephaly. Broad gyri along the right fronto-parietal cortex suggestive of lissencephaly. Absence of septum pellucidum is also noted.

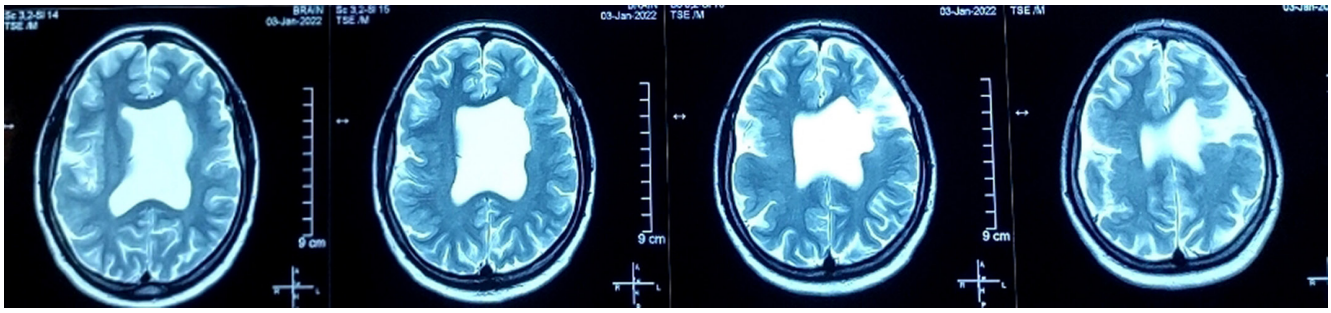

FIGURE 2 T2 image in axial plane showing hyperintense cortical cleft.

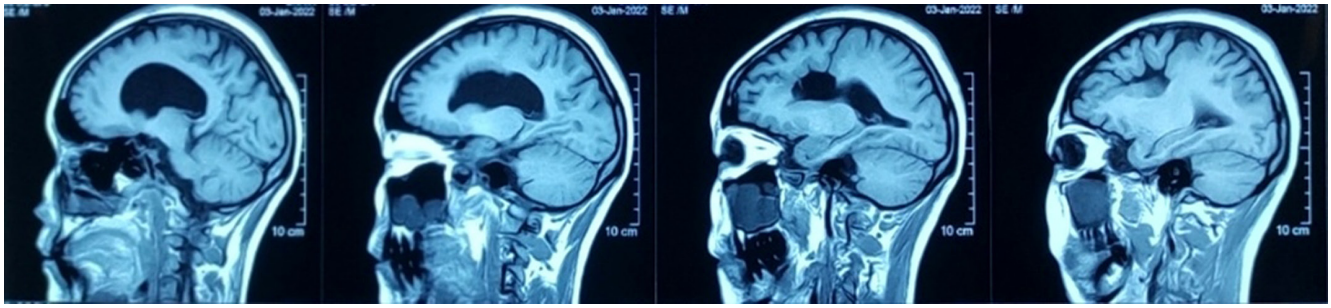

FIGURE 3 T1 image in sagittal plane showing lateral ventricular fusion.

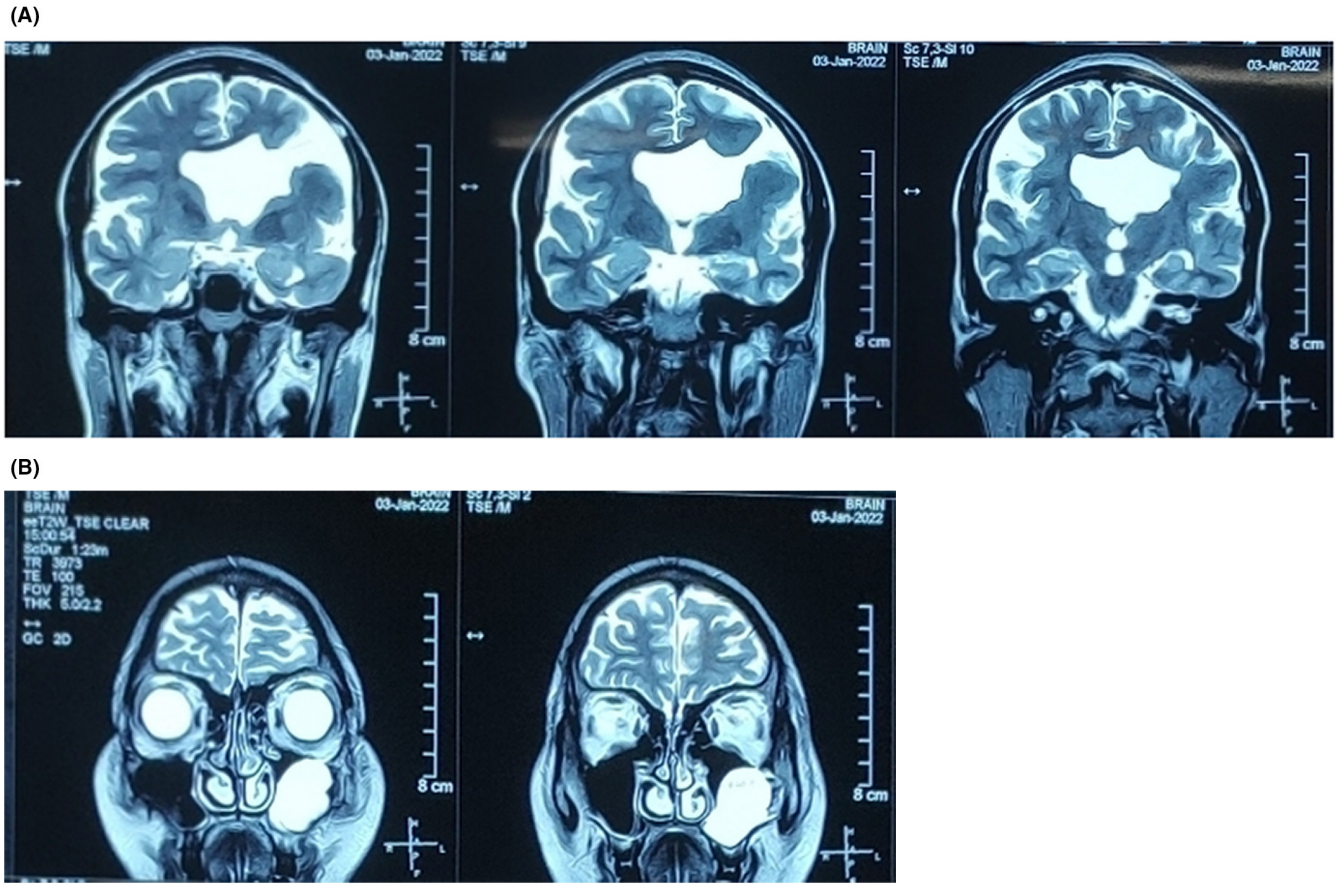


FIGURE 4 T2 coronal view showing (A) hyperintense cortical cleft, and (B) left temporal lobe arachnoid cyst.

and neuronal migration and hence manifest as schizencephaly, lissencephaly, or other forms of cerebral malformation.[6] Another vascular theory suggests that vascular compromise during early embryogenesis can lead to hypoxemia and infarction and thus interfere with the neuronal migration.[8] With evidence of the condition running in families, genetic components are also thought to be operative.[9] Evidences suggest schizencephaly to be a heterogeneous disorder with multiple candidate genes. Mutation in the genes, SHH, ZIC 2, and SIX 3, associated with holoprosencephaly have been found to be linked with schizencephaly thereby demonstrating this disorder as a component of the intricated spectrum of developmental deformities in the ventral forebrain.[3] More recent studies have identified the roles of mutated WDR62 and COL4A1 genes too.[10,11] Nonetheless, schizencephaly has also been seen in cases with no abnormalities in these genes, thereby suggesting the possible roles of various non genetic factors in the complex pathogenesis. Association with in utero Cytomegalovirus infection have also been reported.[12,13] The role of in utero HSV infection in schizencephaly and other spectrum of CNS abnormalities with similar phenotypic traits have also been demonstrated.[14,15]

In more than 50% of the cases, it is seen in association with other congenital abnormalities.[16] Microcephaly, hydrocephalus, septo-optic dysplasia, optic nerve hypoplasia, absent corpus callosum, pachygyria, and polymicrogyria are commonly occurring co existent abnormalities,[5] among which absent septum pellucidum, ventricular deformation, and corpus callosum agenesis are the predominantly encountered ones.[17,18]

In our case, there was absence of septum pellucidum, lissencephaly, lobar holoprosencephaly, and an arachnoid cyst in the left temporal lobe. A theory suggests that the underlying mechanics causing schizencephaly also exerts a dragging force in the meninges thereby leading to formation of arachnoid cyst around the cleft.[19]

The severity of clinical presentation largely correlates with the extent of the underlying anatomical deformity.[17] Motor deficits, epileptic seizures, and mental retardation are mostly seen. Psychotic features such as delusions, hallucinations, and depressive symptoms have also been reported[1] Our patient did not have motor deficits but had frequent episodes of seizures which was present all throughout her pregnancy yet this was the first time she received medical care for it.

Case studies have shown that seizures during pregnancy can have deleterious effects on fetal heart and size,[20] but in our case, even with the non-treated seizure episodes the outcome was a healthy and term baby.

Small unilateral defects can have either no symptoms at all or a milder course with diagnosis being made at fourth or fifth decade or even at later stages of life.[21] In this case, despite the large anatomical deformity and other associated malformations, the diagnosis remained an incidental MRI finding during work up of a seizure during labor in a lady who reportedly had three uneventful pregnancy episodes in the past.

Exact information on age of onset and progression of seizure could not be obtained in our case. But schizencephaly as a cause of adult onset seizure is a rare occurrence and very few cases have been reported in adults.[2,22]

An exceptional feature of our case is the context of the presentation itself: diagnosis made at the time of labor in a woman who was otherwise living a normal lifestyle.

Radiological Imaging is the key diagnostic approach. MRI and CT scans can demonstrate the anatomical location, extent and distinguish among the variants as well.[21] Furthermore, MRI allows distinction between schizencephaly and other acquired lesions along with detection of other co-existing disorders.[18] The visualization of gray matter lining the cleft is pathognomonic of schizencephaly and for this reason MRI is a substantially superior modality of diagnosis. Cleft lined by cortex along with pachygyria and polymicrogyria are commonly appreciated on MRI.[21]

Management is conservative and involves adequate seizure control along with rehabilitation of motor defects and mental retardation.[2] Identification of the epileptogenic foci and surgical resection have been tried with variable outcomes, yet a vigilant approach and accurate resection can yield fulfilling results.[23,24]

## 4 | CONCLUSION

Schizencephaly is rarely diagnosed in adulthood. Apart from rehabilitation and symptomatic management, the need to address patients' traditional beliefs and concerns and ensure treatment adherence was a crucial part in our case. Given the rarity of the disorder in addition to the unusual background of presentation, an absolute need of reporting the case was felt.

### AUTHOR CONTRIBUTIONS

**Kusum Paudel:** Conceptualization; data curation; formal analysis; investigation; methodology; project administration; resources; supervision; visualization; writing – original draft; writing – review and editing. **Tanisha Prasad:** Writing – review and editing. **Prashant Gyawali:** Resources; writing – review and editing. **Gaurav Nepal:** Resources. **Vikash Jaiswal:** Resources; supervision; visualization; writing – review and editing.

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None.

## CONFLICT OF INTEREST STATEMENT

None.

## DATA AVAILABILITY STATEMENT

The data that support the findings of this study are openly available in clinical case reports at https://doi.org/10.22541.

## CONSENT

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

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