International Journal of Scientific Reports
*Dutta G et al. Int J Sci Rep. 2025 Jun;11(6):230-234*
http://www.sci-rep.com
pISSN 2454-2156 | eISSN 2454-2164
DOI: https://dx.doi.org/10.18203/issn.2454-2156.IntJSciRep20251456

**Case Report**

# Open lip schizencephaly: a rare case report on small head in infancy

**Gaurav Dutta, Ekansh Rathoria*, Dakshata Vishnoi, Shaik Aalam, Astha Pandey, Akash Srivastava**

Department of Pediatrics, Hind Institute of Medical Sciences, Sitapur, Uttar Pradesh, India

**Received:** 10 April 2025
**Accepted:** 09 May 2025

***Correspondence:**
Dr. Ekansh Rathoria,
E-mail: rathoriaekansh@yahoo.com



## ABSTRACT

Schizencephaly is a rare congenital brain malformation defined by cerebrospinal fluid-filled clefts that run from the cerebral hemisphere's pial surface to the ventricle's ependymal surface. In this report, we present a 6-month-old male child who presented with complaints of small head size that was not increasing over the last 3 months and inability to control the neck with often increased tone in neck and left side of body, globally delayed developmental milestones, and excessive crying. The magnetic resonance imaging (MRI) of the brain was done, which showed grey-matter lined CSF cleft extending from the pial surface to the ependymal surface of the right lateral ventricle in the right parieto-occipital region, with associated polymicrogyric smooth surface along clefts that was suggestive of right unilateral opened lip schizencephaly. The child was managed conservatively with physiotherapy, syrup baclofen, vitamin-D3, and other multivitamins, and no neuro-surgical intervention was required. This case emphasizes the importance of evaluating children presenting with globally delayed milestones, spasticity, and microcephaly during infancy as soon as possible using MRI, especially in settings with limited resources. Prompt diagnosis, timely intervention, appropriate counseling of the parents, and a multi-disciplinary approach to the management are needed.

**Keywords:** Magnetic resonance imaging, Microcephaly, Nervous system malformations, Schizencephaly, Seizure

## INTRODUCTION

A congenital malformation (CM), often known as a birth defect, is any morphological or functional aberration that is evident at birth and may be genetically inherited or acquired during pregnancy.[1,2] CMs account for 2-3% of all births worldwide.[3] The most common etiology of CMs is idiopathic in nearly 50% of cases, followed by genetic (30-40% cases) and environmental (5-10% cases).[2]

Schizencephaly is a neurologic developmental disorder with a failure in neuronal transmigration.[3] As a result, a cleft that runs from the cerebral hemispheres to the neuroepithelial cells that line the lateral ventricles is created.[3,4] Wilmarth first described schizencephaly in 1887.[5] The global prevalence of schizencephaly is 1.48 per 100,000 births.[5] It is typically sporadic, with only a few familial cases reported, and there is no known gender predisposition.[5] Schizencephaly can be categorized into three types:

***Type I***

The trans-mantle variant, features an aberrant grey matter column that runs from the ependyma to the pia matter without a CSF-containing cleft.[6]

***Type II***

It is known as closed-lip schizencephaly, in which the lips of aberrant grey matter are opposed by CSF-containing cleft.[6]

***Type III***

It is the open-lip variant with non-abutting lining lips of aberrant grey matter and a CSF-containing cleft.[6]

Schizencephaly is diagnosed unilaterally in 63% of cases and bilaterally in 37%.[7] The open lip type is found in 60% of cases of the unilateral variant, primarily in the frontal lobe.[8] Here, we report a rare case of open lip schizencephaly seen at a rural tertiary care center.

## CASE REPORT

### *Chief complaints*

A 6-month-old male infant presented to our pediatrics outpatient department (OPD) at Hind institute of medical sciences, Sitapur, with complaints of small head size that was not increasing over the last 3 months and inability to control the neck with often increased tone in neck and left side of body, globally delayed developmental milestones, and excessive crying.

### *Detailed history*

A detailed history was obtained from the parents. The infant was born of third-degree consanguineous marriage, second in birth order with maternal age of 26 years and paternal age of 27 years at conception. The current pregnancy was confirmed at home by a urine pregnancy test kit at 3 months of gestation. The mother had not registered for antenatal care clinic. There is a history of maternal fever around 2 months of gestation for 2 days that was high grade, continuous, and relieved on taking over-the-counter medication (Tab paracetamol). There was no history of any other drug intake like warfarin, cocaine, or alcohol or radiation exposure during this pregnancy in the mother. There is no history of any attempted abortion, prenatal testing procedure, or maternal trauma. At around thirty-eight weeks of gestation, mother delivered at government hospital through vaginal delivery. As per the documents, the birth weight of the baby was 2500 grams, length 45 cm, and head circumference (HC) 33 cm. The baby did not cry immediately after birth, and neonatal resuscitation for around one minute was carried out, followed by admission to the neonatal intensive care unit (NICU). The baby required oxygen support for the next 24 hours for tachypnea and then was discharged by the third day of life as per documents. The child received exclusive breastfeeding for 6 months as per mother.

### *Examination*

On examination, the child had global delay in the developmental milestones, a grossly small head size with a head circumference of 33 centimeters (cm) that was <3$^{rd}$ percentile according to WHO growth chart 0-5 years boys, and craniofacial disproportion (Figure 1). Anthropometric examination revealed a weight of 6.5 kilograms (kg) (between 3$^{rd}$ and 50$^{th}$ percentile), length 57 cm (<3$^{rd}$ percentile), and mid-upper arm circumference (MUAC) 12.5 cm. The anterior fontanelle was non-bulging and measured 1×1 cm.


**Figure 1 (A and B): General appearance of child showing microcephaly.**

Primitive reflexes like sucking reflexes were intact. There was spasticity in the left upper and lower limbs' muscle tone with a 4/5 power grade (Medical research council grading) and brisk (3+) deep tendon reflexes, while the power on the right upper and lower limbs was 5/5 with normal reflexes. Examination of other systems along with the spine was normal. On ophthalmic examination, pupils were equally normal-sized with normal reactions, lens clear and fundal glow was present in both eyes, and no significant fundal pathology was seen. The infant had microcephaly clinically and was evaluated for the same by investigations.

### *Investigations*

On investigations, hemoglobin was 11.6 grams/deciliter (gm/dl), total leukocyte count was 14300 per cubic millimeter (cmm) with polymorphs 14%, lymphocytes 75%, monocytes 9%, eosinophils 2%, packet cell volume 37.1%, mean corpuscular volume (MCV) 79.8 femtolitres (fl), mean corpuscular hemoglobin (MCH) 25 picogram (pg), MCH concentration 31.3 gm/dl and platelets were 5.22 lakhs/cmm. Red blood cell count was 4.66 million/cmm with a red cell distribution width of 12.4%. The general blood picture was suggestive of microcytic

hypochromic anemia with mild leukocytosis with no haemoparasite or immature cells. The C-reactive protein (0.74 mg/l) was negative. The blood group of the baby was O positive. Liver function tests were within normal limits: Total serum bilirubin 0.40 mg/dl, direct serum bilirubin 0.20 mg/dl, SGOT 49 IU/l, SGPT 47 IU/l, and alkaline phosphatase 492 IU/l. Kidney function tests were within normal limits: blood urea 24 mg/dl, serum creatinine 0.6 mg/dl, and blood urea nitrogen (BUN) 12 mg/dl. Serum electrolytes within normal limits: Serum sodium was 142 mmol/l, serum potassium 4.02 mmol/l and ionic calcium was 1.08 mg/dl. CSF cyto-biochemical analysis was within normal range. Blood and CSF culture sensitivity were sterile. Due to financial constraints, TORCH profile and genetic testing could not be done.

### *MRI*

MRI brain revealed grey-matter lined CSF cleft extending from the pial surface to the ependymal surface of the right lateral ventricle in the right parieto-occipital region, with associated polymicrogyric smooth surface along clefts features suggestive of open-lip schizencephaly (Figure 2). The rest of the brain parenchyma appears normal with normal grey white differentiation. No evidence of abnormal areas of restricted diffusion on DWI or susceptibility blooming on gradient images was noted. The deep nuclei and thalami appear normal in morphology and signal intensity. The basal cisterns, cerebellum, brain stem, and pituitary gland appear normal.

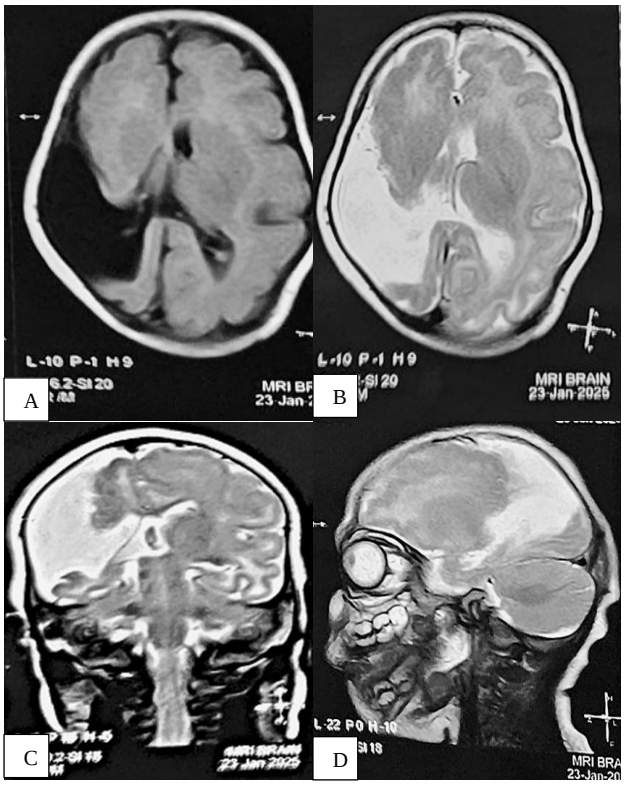


**Figure 2 (A-D): MRI brain suggestive of open-lip schizencephaly.**

### *Management*

The neurosurgery reference was done, and no active neurosurgical intervention was required at present. The patient was managed conservatively with physiotherapy, syrup baclofen, vitamin-D3, and multivitamins. Parents were counseled and explained about poor prognosis. Parents were advised for regular follow-up appointments to monitor and address any potential complications, as well as to seek psychological assistance.

## DISCUSSION

Schizencephaly is an extremely rare congenital brain abnormality.[3.] It is a disorder in the migration of neuronal cells during the second and fifth months of pregnancy.[3]

It is described by cerebrospinal fluid-filled clefts that run from the cerebral hemisphere's pial surface to the ventricle's ependymal surface.[9] Three clinical variants, namely trans-mantle variant, open lip, and closed lip, have been described.[3] It can be bilateral or unilateral, symmetrical or asymmetrical, involving any part of the brain.[9] The most commonly involved areas are the parietal and frontal lobes, particularly the sylvian fissure region.[9] Our case presented with right unilateral open-lip schizencephaly (Figure 2).

Other CMs like agenesis of the septum pellucidum and corpus callosum, polymicrogyria (an excessive number of small, partly fused gyri), pachygyria (unusually thick convolutions of the cerebral cortex), heterotopias (ectopic gray matter), septo-optic dysplasia, hypoplasia of optic nerve and enlarged ventricles are reported in 50-90% of cases of schizencephaly.[10] Our patient had none of the aforementioned associations except for associated polymicrogyric smooth surface along clefts.

The pathophysiology of schizencephaly is unclear.[9] Nonetheless, the inhibition of neuronal migration is thought to be the cause.[9] Neuroblasts move from the germinal matrix to the cerebral cortex area between the seventh and eighth weeks of gestation, following the radially orientated glia cells.[9] Any major vascular injury at this critical site may cause neuroblast migration to fail, which would result in the creation of a cleft.[9]

Although the exact cause of this condition is unknown, a number of speculations have linked it to teratogenic substances like alcohol, warfarin, or cocaine, viral infections like cytomegalovirus (CMV), Zika virus, and herpes simplex virus (HSV), foetal brain ischemic injury, stroke in-utero, hypoxia in 8$^{th}$ week of pregnancy, abortion trial, prenatal testing procedures like amniocentesis, or chorionic villus biopsy, trauma to mother, and young maternal age.[3,9,11,12]

There have also been some documented familial cases of schizencephaly, albeit the underlying genetic reason for these cases is still unclear.[13] Schizencephaly may be

caused by certain genetic mutations, according to certain studies.[11] The following are the primary genes found in this context: Mutations in the EMX2-germline, SIX3, SHH, and COL4A1 genes.[11]

The possible aetiology in our case can be due to intrauterine hypoxic insult or intrauterine infection (as the mother had a fever at 2 months of gestation), which could have led to the disruption of the germinal matrix vascular supply and further schizencephaly.

Depending on the extent of neuronal involvement, different clinical symptoms occur, which vary from asymptomatic to severely neurologically disabled patients.[9] Common symptoms include motor impairment (90% cases) varying from hemiparesis to tetraplegia, neurocognitive impairment (77.5% cases), epilepsy (67.5% cases), seizures, delayed milestones, intellectual disability, and microcephaly.[9] Closed lip schizencephaly patients tend to have lesser neurological deficits than open lip schizencephaly patients.[9] Hemiparesis and motor delay are symptoms of closed lip schizencephaly, while hydrocephalus and seizures are symptoms of open lip schizencephaly.[14]

Our patient presented with a small head size that was not increasing over the last 3 months that was <3$^{rd}$ percentile at 6 months age according to WHO growth chart 0-5 years boys (microcephaly), not able to hold his neck at 6 months of age with often increased tone in neck and left side of body (spasticity), delayed developmental milestones, and excessive crying.

The preferred neuroimaging method of choice for schizencephaly assessment is MRI, which shows the pathognomonic finding of a grey matter-lined cleft in the cerebral cortex.[9] MRI is more sensitive than computerised tomography (CT) scans and can easily distinguish between grey and white matter.[9] However, an in-utero diagnosis can also be made based on an antenatal ultrasound scan.[15] In our case, MRI brain revealed grey-matter-lined CSF cleft extending from the pial surface to the ependymal surface of the right lateral ventricle in the right parieto-occipital region, with associated polymicrogyric smooth surface along clefts that was suggestive of open-lip schizencephaly (Figure 2).

Schizencephaly is primarily managed conservatively.[9] It entails using antiepileptic medications to manage seizures in addition to rehabilitation for mental retardation and motor impairments.[9] When hydrocephalus and intracranial hypertension occur together, surgery is necessary; in these cases, a ventricular shunt is usually inserted.[9] Our patient was managed conservatively with physiotherapy, syrup baclofen, vitamin-D3, and multivitamins, and the parents were counseled for its poor prognosis. The differential diagnosis of schizencephaly includes acquired cysts, hydrocephalus, holoprosencephaly, focal cortical dysplasia, grey matter heterotopia, and porencephaly.[5]

## CONCLUSION

Although schizencephaly is a very rare congenital malformation, it should be considered a possibility during infancy in children presenting with delayed milestones, spasticity, and microcephaly, as in this case, which was diagnosed using MRI and managed conservatively. This case highlights the necessity for prompt radiological investigation with MRI in the evaluation of these cases, particularly in resource-limited settings where it is not routinely done. Parents should be counseled for the poor prognosis. To prevent such unanticipated birth defects that have a significant psychological impact on the family, proper prenatal follow-up is strongly recommended.

*Funding: No funding sources*
*Conflict of interest: None declared*
*Ethical approval: Not required*

## REFERENCES

1. Congenital disorders. Who.int. Available at: https://www.who.int/news-room/fact-sheets/detail/birth-defects/. Accessed on 25 February 2025
2. Shrestha S, Shrestha A. Prevalence of congenital malformations among babies delivered at a tertiary care hospital. J Nepal Med Associat. 2020;58(225):310.
3. Dirani FA, Makiye ZA, Nemer ZH. Case Report: Giant Bilateral Open Lip Schizencephaly in Newborn Girl. Clin Image Case Rep J. 2024;6(2):379.
4. Hakimi T, Rahimi HM, Jawed MA. Unilateral open lip Schizencephaly. J Pediatr Surg Case Rep. 2021;73:102011.
5. Veerapaneni P, Veerapaneni KD, Yadala S. Schizencephaly. In: StatPearls. Treasure Island (FL): StatPearls Publishing. 2025.
6. Griffiths PD. Schizencephaly revisited. Neuroradiology. 2018;60:945-60.
7. Pooh RK, Matsuzawa N, Machida M, Nakamura T, Chiyo H. Neurosonographic Approach to Malformations of Cortical Development. Donald School J Ultrasound Obstetr Gynecol. 2021;15(2):179-87.
8. Khayi F, Bennaoui F, El Idrissi N. Fmr Maoulainine. Open Cleft Schizencephaly: Rare Etiology of Macrocrania in Newborns: About A Case. Sch J Med Case Rep. 2023;5:922-4.
9. Okunola OP, Olaniyi OK, Oronsaye EJ. Schizencephaly: a case report and review of literature. Open J Pediatr. 2022;12(5):742-8.
10. Laasri K, Naggar A, El Houss S, Halfi MI, Taoursa F, Kettani NE, et al. Schizencephaly: A rare cause of late-onset epilepsy in an adult. Radiol Case Rep. 2023;18(11):3861-4.
11. Khadka C, Gupta U, Bhandari P, Pandey P, Paudel S. A case of closed-lip schizencephaly with absent septum pellucidum in an adult presenting with

seizure disorder. Clin Case Reports. 2023;11(6):e7536.
12. Rathoria E, Bansal U, Saxena A. Zika Virus Disease: A Review. Child Today. 2021;4(4):19-24.
13. Doğan E. The evaluation of unilateral closed-lip schizencephaly on complex MRI sequences. Rom J Neurol. 2021;20:250-4.
14. Hakimi T, Qasem KM. Bilateral open lip schizencephaly. Ann Med Surg. 2022;73:103204.
15. Ugboma EW, Agi CE. Schizencephaly: A case report and review of literature. Nigerian Postgrad Med J. 2016;23(1):38-40.

**Cite this article as:** Dutta G, Rathoria E, Vishnoi D, Aalam S, Pandey A, Srivastava A. Open lip schizencephaly: a rare case report on small head in infancy. Int J Sci Rep 2025;11(6):230-4.