

## Case Report

## Bilateral open lip schizencephaly

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## ABSTRACT

Schizencephaly is a central nervous system (CNS) developmental disorder characterized by abnormal cleft extending from the lateral ventricles to the cerebral cortex. Clinically, it occurs as *trans*-mantle, closed lip and open lip types which may be unilateral or bilateral. The exact cause of schizencephaly is not known but genetic disorders, exposure to teratogens, viral infections and maternal age are implicated. We present a case of bilateral open lip schizencephaly with some degrees of neurological disorders caused by increased intra-cranial pressure (ICP) due to ventriculomegaly. We applied ventriculo-peritoneal shunt (V-P shunt) to the patient with considerable improvement after post-operative follow-up.

## 1. Introduction and importance

Schizencephaly is a rare congenital anomaly characterized by the failure of neuronal migration and resultant cleft extending from cerebral hemispheres to neuroepithelial cells lining the lateral ventricles [1,2]. The prevalence of schizencephaly is 1.48/100 000 live births which is more common in younger mothers [3]. The etiology is not well understood but teratogens, viruses mostly cytomegalovirus, alcohol and narcotics, hypoxia, maternal age and trauma may be involved [4–6]. Most of the schizencephaly cases are sporadic and non-familial with no any known causative etiology [14]. Computerized tomography scan and Magnetic resonance imaging (CT-Scan and MRI) are the modalities of choice for diagnosis but MRI is superior for better differentiation between gray and white matters [7,8]. Treatment is supportive and surgery is needed if there is hydrocephalus [7].

## 2. Case presentation

We present a case of 30 months old child brought by her parents to our department suffering from head enlargement associated with internal strabismus and lower limbs low degree weakness. The patient was born to a consanguineous couple with no familial history for the mentioned problem. According to the patient parents information, the child head started to expand with the eye balls moving toward midline at the age of 4 months. They took their child to the local clinic and received appropriate advice by the relevant physician. In the next visit for the purpose of definite treatment, the patient was referred to another

physician who had private practice in the downtown. Here the patient was taken under regular conservative treatment with fixed intervals. This process lasted for two years with no positive result but worsening the condition, so the patient was referred for specialized treatment to our hospital pediatric surgery department. Our team evaluated the patient by performing the relevant clinical examinations. Patient was weighting 11kg and with physical exam her head was enlarged with the internal strabismus and lower limbs weakness but all reflexes still remained intact and no history of seizures (common in schizencephaly). Our team carried out all biochemical tests along with brain CT-Scan. Biochemical tests were normal, but CT-Scan showed bilateral cysts separating by a cleft, resembling bilateral open lip schizencephaly (Fig. 1). As there was considerable ventriculomegaly, our team advised VP-Shunt for the purpose of decreasing ICP. We treated the patient by placement of VP-Shunt with no any intra-operative and post-operative complications (Figs. 2–6). The operation ended uneventfully.

## 3. Discussion

Schizencephaly is uncommon cortical anomaly resulting from split brain due to the cleft extending from pia matter to the ependymal cell of lateral ventricles. The condition was first described by Yakovlev and Wadsworth in 1946 as the infolding of gray matter along the clefts which are located in the area of primary fissure and associated with other CNS anomalies [15]. Closed lip type of schizencephaly is characterized by inter-connected gray mater lined lips whereas the open lip type is characterized by open lips with a CSF filled cleft extending to the lateral

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ventricles [9]. Our case was bilateral open lip type of schizencephaly.

The cause is unknown but failure of neuronal migration along the radially oriented glial cells in the brain cortex due to any type of insult and resultant hypoxemia with infarction in the 8th week of gestation is hypothesized [9,10]. Additionally viral infection (cytomegalovirus) and more recently Zika virus, attempted abortion, hypoxia, maternal age and trauma are also involved [6,11,16].

Signs and symptoms depend on the clinical type. Closed lip schizencephaly is manifested by hemiparesis and motor delay whereas open lip schizencephaly manifested by hydrocephalus and seizures [12]. Our patient was complaining of diplegia, internal strabismus and hydrocephalus but not history of seizures (rare).

Definite diagnosis is given by history, signs, symptoms and imaging modalities (CT-Scan & MRI). Treatment is mostly supportive focusing on seizures control and rehabilitation of physical and mental status. Surgery is not needed but if there are signs and symptoms of increased ICP especially hydrocephalus, V-P Shunt application is mandatory [13]. Conditions such as focal cortical dysplasia with cleft in the cortex but not extending to the ventricles, gray matter heterotopia seen as a linear cleft but periventricular gray matter generally bulge into the ventricles and procencephaly extension from the cortex to ventricles but lined by



Fig. 2. Prepared right temporo-Parietal area for incision.

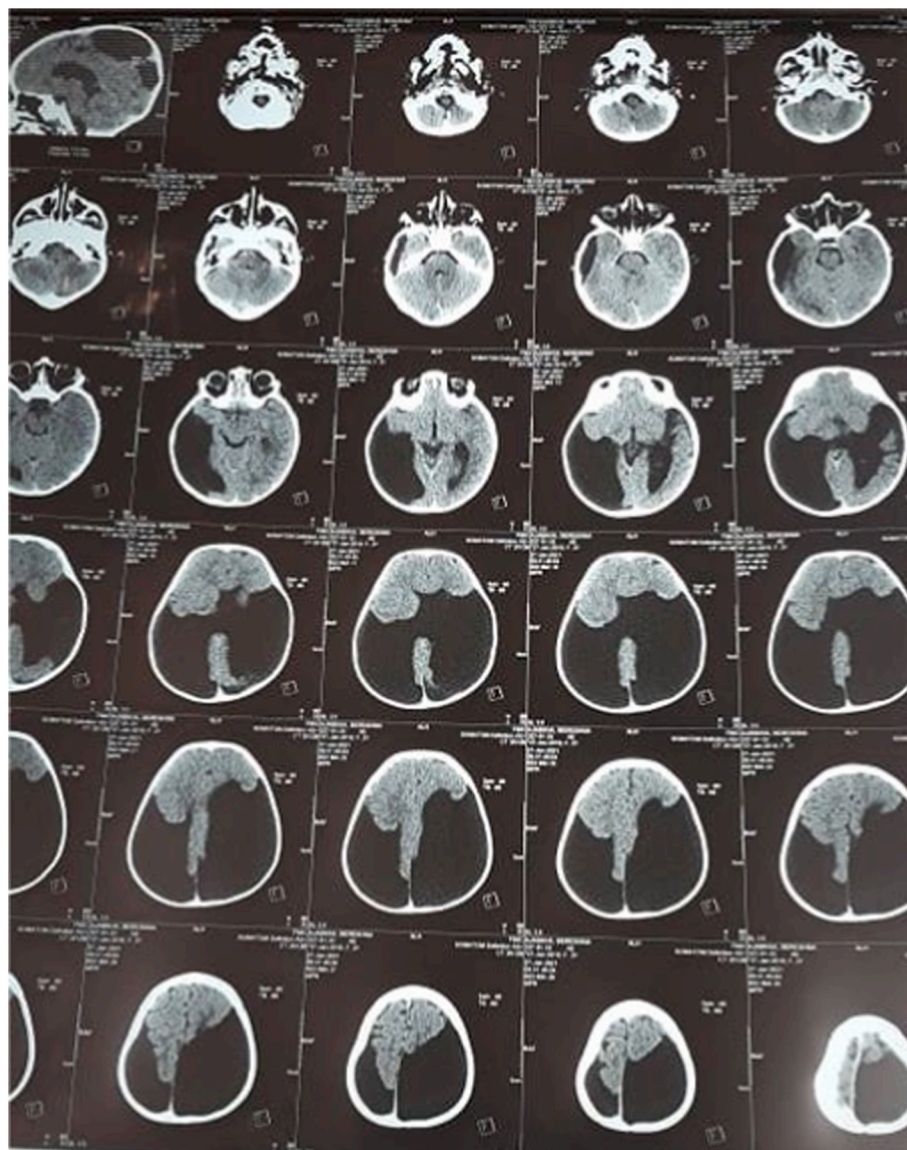


Fig. 1. CT-Scan image, showing cleft extending from Pia mater to the lateral ventricles.



Fig. 3. Scalp incision.



Fig. 4. Skull bur hole using drill.

gliotic white matter are all that should be considered in differential diagnosis of schizencephaly [17,18]. As our patient was suffering from hydrocephalus, we applied V-P Shunt and the operation ended uneventfully. Six months following discharge from the hospital, the patient's mentioned complains considerably relieved (patient's condition description by her father through phone contact).

#### 4. Conclusion

Schizencephaly is still prevalent in developing nations and on time management will improve the patient condition and prevent disability. Our case was from a rurally lived illiterate family and low socioeconomic condition with no direct access to primary healthcare system and referral to specialized center. We assume that antenatal viral infections



Fig. 5. V-P Shunt application (insertion process of ventricular end of shunt along with guider).

(ante-natal viral screening is not done routinely in rural areas), addiction (not alcohol but narcotics which are common in some families where they cultivate opium in their farms and become addicted), maternal age and sometimes (maternal trauma due to violence) may be implicated as causes. Further research should be carried out to know more about the causes of CNS anomalies in developing nations.

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No ethical approval was necessary.

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Fig. 6. Wound closure in the neck and head.

#### Author contribution

Turyalai Hakimi (TH) performed the procedure and conceptualized the manuscript. TH and Khalid M, Qasem (KMQ) designed the study. TH and KMQ contributed for the diagnosis and treatment. Both TH and KMQ wrote the original draft and edited the manuscript with the supervision of the entire study process. Both authors read and approved the final manuscript.

#### Registration of research studies

1. Name of the registry: None.
2. Unique Identifying number or registration ID: None.
3. Hyperlink to your specific registration (must be publicly accessible and will be checked): None.

#### Guarantor

Dr. Turyalai Hakimi.

#### 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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#### Authorship

All authors attest that they meet the current ICMJE criteria for

authorship.

#### Declaration of competing interest

The authors declare that they have no known competing financial interests of personal relationships that could have appeared to influence the work reported in this paper.

#### References

- [1] A. Tanwir, S. Bukhari, M.S. Shamim, Frontoethmoidal Encephalocele Presenting in Concert with Schizencephaly, *Surgical neurology international*, 2018 [PubMed PMID: 30603230].
- [2] P.C. Hung, H.S. Wang, M.L. Chou, K.L. Lin, M.Y. Hsieh, I.J. Chou, A.M. Wong, Schizencephaly in children: a single medical center retrospective study, *Pediatr. neonatol.* (2018 Dec) [PubMed PMID: 29371079].
- [3] Howe DT, Rankin J, Draper ES, Schizencephaly prevalence, prenatal diagnosis and clues to etiology: a register-based study. *Ultrasound Obstet.*, [PubMed PMID: 21647999].
- [4] P.D. Griffiths, Schizencephaly Revisited, *Neuroradiology.*, 2018 Sep [PubMed PMID: 30027296].
- [5] J.C. Gonzalez, K. Singhapakdi, A.M. Martino, B.H. Rimawi, R. Bhat, Unilateral open-lip schizencephaly with tonsillar herniation in a preterm infant, *J. Pediatr. Neurosci.* (2019 Oct-Dec) [PubMed PMID: 31908665].
- [6] K.A. Dies, A. Bodell, F.M. Hisama, C.Y. Guo, B. Barry, B.S. Chang, A.J. Barkovich, C.A. Walsh, Schizencephaly: association with young maternal age, alcohol use, and lack of prenatal care, *J. Child Neurol.* (2013 Feb) [PubMed PMID: 23266945].
- [7] A. Halabuda, L. Klasa, S. Kwiatkowski, L. Wyrobek, O. Milczarek, A. Gergont, Schizencephaly-diagnostics and clinical dilemmas, *Child's Nerv. Syst.: ChNS: off. j. Intern. Soc. Pediatr. Neurosurg.* (2015 Apr) [PubMed PMID: 25690450].
- [8] V. Kamble, A.M. Lahoti, A. Dhok, A. Taori, N. Pajnigara, A rare case of schizencephaly in an adult with late presentation, *J. Fam. Med. Prim. Care* (2017 Apr-Jun) [PubMed PMID: 29302567].
- [9] H.B. Sarnat, Role of human fetal ependyma, *Pediatr. Neurol.* 8 (1992) 163–178 [PubMed] [Google Scholar].
- [10] D. Denis, J.F. Chateil, M. Brun, O. Brissaud, D. Lacombe, D. Fontan, et al., Schizencephaly: clinical and imaging features in 30 infantile cases, *Brain Dev* 22 (2000) 475–483 [PubMed] [Google Scholar].
- [11] T. Granata, L. Farina, A. Faiella, R. Cardini, L. D'Incerti, E. Boncinelli, et al., Familial schizencephaly associated with EMX2 mutation, *Neurology* 48 (1997) 1403–1406 [PubMed] [Google Scholar].
- [12] I. Kopyta, E. Jamroz, E. Marszal, E. Kluczevska, Schizencephaly—clinical and radiological presentation of pediatric patients, *Wiad. Lek.* 59 (2006) 471–476 [PubMed] [Google Scholar].
- [13] A. Halabuda, L. Klasa, S. Kwiatkowski, L. Wyrobek, O. Milczarek, A. Gergont, Schizencephaly-diagnostics and clinical dilemmas, *Child. Nerv. Syst.* 31 (2015) 551–556 [PMC free article] [PubMed] [Google Scholar].
- [14] A.C. Hilburger, J.K. Willis, E. Bouldin, A. Henderson-Tilton, Familial schizencephaly, *Brain Dev.* 15 (3) (1993) 234–236, [https://doi.org/10.1016/0387-7604\(93\)90072-G](https://doi.org/10.1016/0387-7604(93)90072-G) [PubMed] [CrossRef] [Google Scholar].
- [15] H.B. Sarnat, Role of human fetal ependyma, *Pediatr. Neurol.* 8 (1992) 163–178 [PubMed] [Google Scholar].
- [16] M.A. Honein, A.L. Dawson, E.E. Petersen, Birth defects among fetuses and infants of US women with evidence of possible Zika virus infection during pregnancy, *JAMA* 317 (1) (2017) 59–68, <https://doi.org/10.1001/jama.2016.19006> [PubMed] [CrossRef] [Google Scholar].
- [17] T. Harada, T. Uegaki, K. Arata, T. Tsunetou, F. Taniguchi, T. Harada, Schizencephaly and porencephaly due to fetal intracranial hemorrhage: a report of two cases, *Yonago Acta Med.* 60 (4) (2017 Dec) 241–245 [PMC free article] [PubMed].
- [18] U. Senol, K. Karaali, B. Aktekin, S. Yilmaz, T. Sindel, Dizygotic twins with schizencephaly and focal cortical dysplasia, *AJNR Am. J. Neuroradiol.* 21 (8) (2000 Sep) 1520–1521 [PMC free article] [PubMed].