Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Medicine

Open Cleft Schizencephaly: Rare Etiology of Macrocrania in Newborns: About A Case

FZ. Khayi[1*], F. Bennaoui[2], NS EL Idrissi[3], FMR Maoulainine[4]

[1]Neonatal Intensive Care Unit; Mother-Child Hospital; University Hospital Mohammed VI Marrakesh, Morocco
Research Laboratory: Children, Health and Development, FMPM, Cadi Ayad University, Marrakesh, Morocco
[2]Associate Professor; Neonatal Intensive Care Unit, Mother and Child Hospital, Mohammed VI University Hospital, Marrakesh, Morocco
Childhood Health and Development Research Team, Marrakech Medical School, Cadi Ayyad University, Marrakesh, Morocco
[3]Senior Professor; Neonatal Intensive Care Unit, Mother and Child Hospital, Mohammed VI University Hospital, Marrakesh, Morocco
Childhood Health and Development Research Team, Marrakech Medical School, Cadi Ayyad University, Marrakesh, Morocco
[4]Head of department of Neonatal Intensive Care Unit, Mother and Child Hospital, Mohammed VI University Hospital, Marrakesh, Morocco
Childhood Health and Development Research Team, Marrakech Medical School, Cadi Ayyad University, Marrakesh, Morocco

DOI: 10.36347/sjmcr.2023.v11i05.042 | **Received:** 02.04.2023 | **Accepted:** 09.05.2023 | **Published:** 13.05.2023

***Corresponding author:** Fz. Khayi
Neonatal Intensive Care Unit; Mother-Child Hospital; University Hospital Mohammed VI Marrakesh, Morocco

Abstract **Case Report**

Schizencephaly is a rare anomaly of embryonic development characterized by the presence of linear clefts containing cerebrospinal fluid and lined with dysplastic gray matter; which extends from the lateral ventricle to the outer surface of the cortex. The schizencephalic cleft can be open or closed Established etiologies include in-utero infections, young maternal age, abuse of drugs and toxic products. The clinic is dominated by motor impairment, mild mental retardation and epileptic seizures. We report the case of a newborn with macrocrania related to unilateral open cleft schizencephaly.

Keywords: Agenesis of the corpus callosum; Dandy Walker malformation; macrocrania; newborns**;** open cleft.

Copyright © 2023 The Author(s): This is an open-access article distributed under the terms of the Creative Commons Attribution **4.0 International License (CC BY-NC 4.0)** which permits unrestricted use, distribution, and reproduction in any medium for non-commercial use provided the original author and source are credited.

INTRODUCTION

Schizencephaly is a rare congenital malformation of cerebral cortical development characterized by a cleft extending from the surface of the pia mater to the cerebral ventricles. In 1946 Yakovlev and Wadsworth found the presence of clefts in the brain wall during an autopsy series, which they called schizencephaly [1]. Two anatomical types of schizencephaly are described: Type I (closed cleft walls) and Type II (open cleft walls with coexisting "hydrocephalus"). The edges of the cleft are lined with heterotropic and dysplastic gray matter [2]. We report the case of a newborn who was hospitalized for macrocrania with a weak sucking reflex since, related to unilateral open cleft schizencephaly. This clinical observation makes it possible to highlight the clinical and paraclinical aspects of this rare entity in the newborn whose diagnosis can be made prenatally.

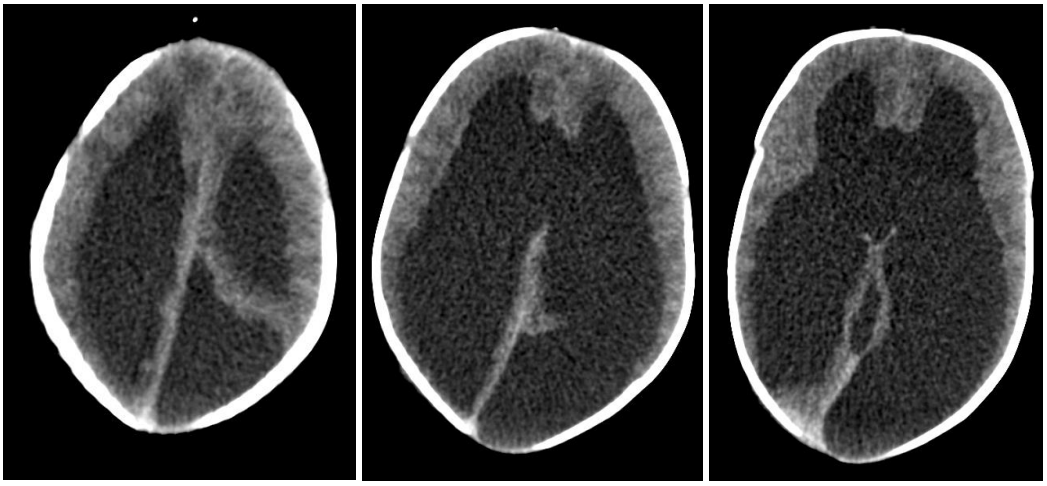
CLINICAL OBSERVATION

Newborn male from a 30-year-old mother; no particular pathological history; youngest of three siblings; from an unrelated marriage; the pregnancy was monofetal followed estimated at 37 weeks + 2 days according to the date of the last menstrual period and 39 weeks + 4 days according to the FARR score; the delivery was by caesarean section for hydrocephalus in labour; with an APGAR of 08/10th reduced to 10/10th at the 5th minute; the infectious anamnesis was negative.

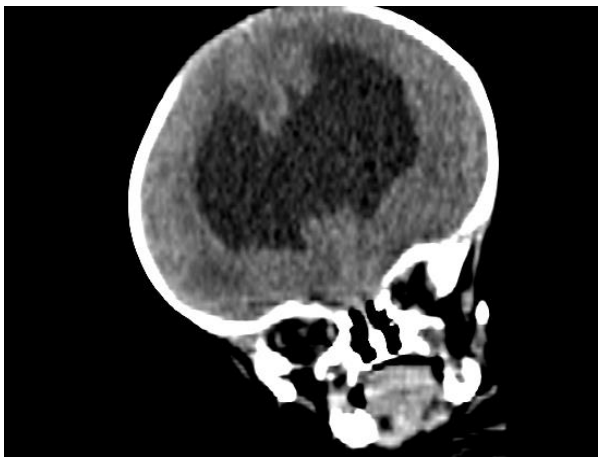
At 17 hours of life, the newborn having presented a regression of the sucking reflex for which he was hospitalized in neonatal intensive care unit, the examination on admission having objectified a reactive pink newborn gesticulating spontaneously; heart rate at 122 beats/min; eupneic at 33 cycles/min arterial oxygen saturation: 93% in the open air a macrocranium with a cranial perimeter at 48.4cm (>97 percentiles); weight: 3kg800 (>75 percentiles); height: 50 cm (50 percentiles).

During his hospitalization, a transfantanel ultrasound was performed showing an aspect of schizencephaly with major triventricular hydrocephalus associated with a cystic formation of the posterior cerebral fossa; Complementary CT scan was done showing left parietal open cleft schizencephaly associated with Dandy Walker malformation with agenesis of the corpus callosum.

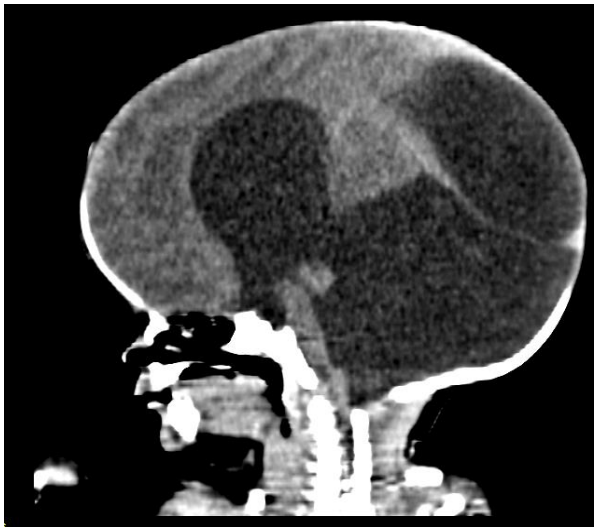
The biological assessment was normal; the newborn was sent to the neurosurgery department for surgical management of his major hydrocephalus.



Cerebral CT without injection of the contrast agents in axial section objectively Dilatation of the lateral ventricles more marked at the level of the occipital horns (colpocephaly) with laminated aspect of the cerebral cortex opposite. Demonstration of a left parietal parenchymal cleft bordered by a cortical ribbon, communicating the subarachnoid spaces with the ipsilateral VL crossroads in relation to a Schizencephaly with open slot



Cerebral CT in coronal slice Dilation moderate and ascent of the V3 with separation of the front horns of the VLs achieving the "bull's horn" appearance.



Brain CT in sagittal section objectifying Vermian agenesis with cystic formation of the posterior cranial fossa seeming to communicate with the V4 and ascent of the tent of the cerebellum and the torcular.

DISCUSSION

Schizencephaly is a cleft of gray matter filled with cerebrospinal fluid and extends from the lateral ventricle to the outer surface of the cortex. There are two clinical types (open and closed cleft) which can be unilateral or bilateral. In the unilateral variety, the open slit type occurs in 60% of cases and mainly in the frontal lobe [2].

Schizencephaly is an anomaly of cortical development occurring during pregnancy [3]. Its incidence worldwide has been estimated at 1.5 in 1,000,000 live births and 1 in 1,650 in children with epilepsy. The majority of cases are believed to be sporadic. No gender predilection has been noted [4]. However, only one study by Stopa et al. finds a male predominance [4]. A study by Howe et al. found that schizencephaly was more common in infants born to younger mothers [6]. The incidence and prevalence in Morocco and Africa are unknown.

Although no specific cause has been identified, various hypotheses have been made. It is thought to be due to abnormal neuronal migration or localized ischemia [5, 7, 8]. Other causes are the expression of genetic factors such as the mutant EMX2 gene. These expressed genetic factors are thought to damage the periventricular germinal matrix, impairing cell migration at 6–7 weeks of intrauterine growth [7]. Sarnat and Curatolo in their research, described it as an extreme form of true porencephaly due to the ischemic theory [8].

Schizencephaly is often associated with other congenital anomalies in 50-90% of cases [8] such as agenesis of the septum pellucidum and corpus callosum, polymicrogyria (excessive number of partially fused small gyri), pachygyria (convolutions unusually thick cerebral cortex), heterotopias (ectopic gray matter), septo-optic dysplasia, and optic nerve hypoplasia. This patient presented with agenesis of the corpus callosum and a Dandy Walker malformation in addition to the open left parietal cleft.

The diagnosis of schizencephaly is made by neuroimaging; computed tomography or magnetic resonance imaging, the latter being the most effective [3]; for our patient the diagnosis was made by computed tomography.

Management of schizencephaly is primarily conservative, including seizure control as well as management of motor deficit and intellectual problems. Surgery is indicated when there are signs of increased intracranial pressure, primarily due to hydrocephalus [9] Surgical management is associated with a risk of complications such as postoperative bleeding, empyema, meningitis, hydrocephalus, and distention of paracerebral fluid spaces forming hygromas or subdural hygrohematomas [10].

CONCLUSION

This case of schizencephaly revealed by macrocrania demonstrates the interest of prenatal consultations. This also makes it possible to discuss new prevention strategies and an improvement in the supply of care. Because developmental anomalies occurring during pregnancy are a real public health problem in our African countries. They are the cause of significant maternal and fetal mortality, physical disability with serious consequences on the quality of life and society.

REFERENCE

1. Yakovlev, P. I., & Wadsworth, R. C. (1946). Schizencephalies: a study of the congenital clefts in the cerebral mantle: II. Clefts with hydrocephalus and lips separated. *Journal of Neuropathology & Experimental Neurology*, *5*(3), 169-206.
2. Hakimi, T., Rahimi, H. M., & Jawed, M. A. (2021). Unilateral Open Lip Schizencephaly. *Journal of Pediatric Surgery Case Reports*, *73*, 102011.
3. Apo, OO, Gnigone, PM, Mapaga, JN, Ongoly, S., Eteno, MN, Matsalou, GM, ... & Ndouongo, PK (2022). Open slit schizencephaly: about a case. *HEALTH SCIENCES AND DISEASE* , *23* (4).
4. Ugboma, E. W., & Agi, C. E. (2016). Schizencephaly: A case report and review of literature. *Nigerian Postgraduate Medical Journal*, *23*(1), 38-40.
5. Stopa, J., Kucharska-Miąsik, I., Dziurzyńska-Białek, E., Kostkiewicz, A., Solińska, A., Zając-Mnich, M., ... & Samojedny, A. (2014). Diagnostic imaging and problems of schizencephaly. *Polish Journal of Radiology*, *79*, 444.
6. Howe, D. T., Rankin, J., & Draper, E. S. (2012). Schizencephaly prevalence, prenatal diagnosis and clues to etiology: a register-based study. *Ultrasound in obstetrics & gynecology*, *39*(1), 75-82.
7. Tietjen, I., Erdogan, F., Currier, S., Apse, K., Chang, B. S., Hill, R. S., ... & Walsh, C. A. (2005). EMX2-independent familial schizencephaly: Clinical and genetic analyses. *American Journal of Medical Genetics Part A*, *135*(2), 166-170.
8. Sarnat, H.B., & Curatolo, P., (2007). Malformations of the nervous system. 1Sted. Edinburgh: *Elsevier Publishers, Science Health Division*. p. 235.
9. Halabuda, A., Klasa, L., Kwiatkowski, S., Wyrobek, L., Milczarek, O., & Gergont, A. (2015). Schizencephaly-diagnosis and clinical dilemmas. *Childs Nerv Syst*, 31(4):551–6
10. Siti, B. C., Zulkifli, M. M., Yusoff, S. S. M., Muhamad, R., & Ahmad, T. M. (2020). A Rare Case of an Infant with Left Hemiparesis: A Case Report of Bilateral Open-lip Schizencephaly. *Malaysian family physician: the official journal of the Academy of Family Physicians of Malaysia*, *15*(3), 90.