



## Occipital encephalocele : about a case

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### Introduction

Encephalocele is a malformation due to a neural tube closure defect resulting in herniation of brain tissue and/or meninges through this congenital skull defect. The size of the encephalocele varies from a few centimeters to a huge mass called a "giant encephalocele". The contents usually consist of degenerated neural tissue, meninges and a cystic part. A newborn at H0 of life, female, born to consanguineous parents, from a sibling of 2. Examination had revealed a weight of 3500g, a head circumference of 35cm with a non bulging anterior fontanel. She had a huge renal occipital mass of 23 x 28 cm and a height of 14 cm. The transillumination was positive. All our work was reported in accordance with the SCARE criteria and guidelines (20).

### Case report:

A 4-month-old newborn, born to consanguineous parents, from a 36-year-old IIG and IIP mother, had reported no pathology during pregnancy but had reported to us the use of ventolin first trimester medication. She had attended 3 prenatal counseling sessions, the first two in the second trimester of pregnancy and the last one in the third trimester. She had received folic acid during the first trimester. A morphological ultrasound at 34 weeks of age revealed a large meningocele with extra cerebral development communicating with the posterior cerebral fossa with cerebellar and vermian atrophy and a supratentorial ventriculomegaly (Figure 1). An emergency cesarean section was performed as soon as the patient went into labor. It resulted in the birth of a female newborn, weighing 2800 g, APGAR score 8/10. At birth, the clinical examination revealed a weight of 2800 g, a head circumference of 33 cm with a non bulging anterior fontanel. The infant appeared active. The neurological examination was normal. She had a huge renal occipital mass measuring 43 x 25 cm and 15 cm in size with positive transillumination (Figure 2 and 3). The rest of the external morphological examination was normal.

### Discussion

Encephaloceles are defined as herniation of brain tissue and/or meninges out of the cranium through a congenital bony defect due to a defect in closure of the cranial portion of the neural tube [2]. Encephaloceles, spina bifida, and anencephalies are commonly grouped under the term congenital neural tube defects (CNTDs) [2,7]. The etiopathogenesis of encephaloceles is still controversial and several theories have been put forward, such as maternal hyperthermia, valproic acid, hypervitaminosis A, vitamin B12 and folic acid deficiency [2,7,8]. An important gene associated with occipital encephalocele is CEP290 (Centrosomal Protein 290) [9]. Occipital encephalocele is frequently associated with neurological disorders,

some infants may be asymptomatic on physical examination [10], but others may present with many different signs and symptoms such as delays in reaching developmental milestones, intellectual disability, learning disabilities, growth retardation, seizures, visual impairment, lack of coordination of voluntary movements (ataxia), hydrocephalus, spastic paraplegia or quadriplegia and microcephaly [11]. The incidence of encephaloceles varies according to geographical area and socioeconomic level [12,13]. In the USA and Western Europe, the incidence of encephalocele is 1 to 3 cases per 10,000 live births [14]. In Canada, the prevalence of encephalocele is 0.4 to 4 cases per 10,000 births (6). In developed countries, neural tube defects are rare largely due to preventive measures. In a multicenter study (11 centers) evaluating preventive folic acid intake between 1987 and 1996, Rosano et al (15) reported a significant decrease in the prevalence of neural tube defects in centers such as Atlanta, England, Wales, Hungary and Japan. Indeed, several studies in sub-Saharan Africa confirm the high frequency of encephaloceles due to low socioeconomic status, consanguinity, poor pregnancy follow-up and young maternal age [2,18]. Efforts to prevent this condition by folic acid supplementation during the periconceptional period and the improvement of national health coverage are elements that explain the decrease in this malformation in Niger [17]. Ndoma et al [19] in the Central African Republic reported 7 cases/year and 4.44 cases/year respectively. Radouani et al. in Morocco [18], reported 68 cases of neural tube closure anomalies including only one case of encephalocele in 4 years. The association of encephalocele with other anomalies such as hydrocephalus, microcephaly, psychomotor and intellectual retardation, Chiari syndrome type II, Dandy-Walker disease is frequent. The occipital location of encephalocele is the most frequent in this study as in the literature; it is also a factor of mortality and morbidity. (3, 5, 8, 16,). Pure occipital localization is the most frequent (1). Parietal localization has a better prognosis than occipital and occipito cervical localizations. (19). Bulky encephalocele (diameter greater than 50mm) (101 cases), is a poor prognostic factor according to Kotil (5). Mixed encephalocele is present in 78.26% of cases (126 cases). It is a meningocele in 16.42% of cases (26 patients), a pure encephalocele without cyst in 6.21% of cases (10 cases). Poor epidermalization and the presence of nervous tissue in the malformation are factors of poor prognosis (3, 5, 9). The age of the child varied from 1 to 4 years with an average age of 8.34 months and 45.96% of the children were admitted before 30 days of age. This average age is higher than in the literature. Antepartum ultrasound in trained hands is the examination of choice in antenatal screening for brain malformations. It allows the detection of a cranial defect with sometimes a hernia of the brain. It shows a mass on the midline of the skull more often in the occipital area than in the frontal area. The average maternal age was 17.2 years in this study. Young maternal age and multiparity are also associated with neural tube defects [11]. The prevalence of encephaloceles, apart from any chromosomal abnormality, is higher in the offspring of mothers younger than 20 years of age compared to those of mothers between 20-24 years and 25-29 years. However, there is no relationship with paternal age (1). A family factor is reported in the occurrence of encephaloceles (11). 67.08% of the cases in this study were consanguineous compared to 41.08% in a previous study (18). Molloy et al (13) report that there is a correlation between the level of folate in the red blood cell in the periconceptional period and neural tube defects. Many authors strongly recommend the implementation of national prevention strategies to reduce the prevalence of NTCAs by taking folic acid in the periconceptional period [6,13]. Surgical repair of encephaloceles can be safely performed when the technical conditions are met. The goal is to achieve a tight, physiologic, and cosmetic closure. The surgical approach varies, and the common pitfall is insufficient closure of the dura resulting in postoperative cerebrospinal fluid (CSF) leakage or pseudomeningocele formation [1,4]. Early surgical treatment prior to the development of significant craniofacial dysmorphism results in satisfactory cosmetic outcomes [6,1,4].

## Conclusion:

Occipital encephalocele is the most common form of encephalocele and manifests as a congenital swelling of various sizes on the occipital bone in the midline. Its discovery in a patient requires the search for associated malformations, which in some cases constitutes the amniotic bridges disease complex. The diagnosis is based mainly on the use of imaging techniques. Surgery is the best option for treatment and the appropriate time is between birth and 4 months. The global management starts with the sensitization of the population for a prompt consultation and especially a strengthening of the prevention strategies by the supplementation of folic acid in the periconceptional period, a reinforcement of our technical platform and a multidisciplinary approach for the final correction.

## References

1. Stéphanie A, Black JA, Galvez MA, Schwartz RAJ. Images en anesthésiologie: voies respiratoires (Prise en charge chez un nourrisson atteint d'une encéphalocèle occipitale géante) Anesthésiologie. 2014;120:1504. [[Google Scholar](#)]
2. Singh K, Johnson WMS, Archana R, Kumar A. The prevalence and pattern of neural tube defects and other major congenital malformations of nervous system detected at birth in Barbados. J Anat Soc India. 2016 August;65(Supplement 1):S20–S24. [[Google Scholar](#)]
3. Ghritlaharey RK. Un bref examen de l'encéphalocèle occipitale géante. Journal des neurosciences en milieu rural's entraîner. 2018;9(4):455–6. [[Article PMC gratuit](#)] [[PubMed](#)] [[Google Scholar](#)]
4. Kabré A, Zabsonre DS, Sanou A, Bako Y. The cephaloceles: a clinical, epidemiological and therapeutic study of 50 cases. Neurochirurgie. 2015;61(4):250–4. [[PubMed](#)] [[Google Scholar](#)]
5. Oucheng N, Lauwers F, Gollogly J, Draper L, Joly B, Roux F-E. Frontoethmoidal meningoencephalocele: appraisal of 200 operated cases: Clinical article. J Neurosurg Pediatr. 2010;6(6):541–49. [[PubMed](#)] [[Google Scholar](#)]
6. MalaCards: the human disease database. 2022. MalaCards: the human disease database, Occipital Encephalocele. Accessed on 12 feb 2022. [[Google Scholar](#)]
7. Agarwal A, Chandak AV, Kakani A, Reddy S. A giant occipital encephalocele. APSP J Case Rep. 2010;1(2):16. [[Article PMC gratuit](#)] [[PubMed](#)] [[Google Scholar](#)]
8. Rehman L, Farooq G, Bukhari I. Neurosurgical Interventions for Occipital Encephalocele. Asian J Neurosurg. 2018;13(2):233–237. [[Article PMC gratuit](#)] [[PubMed](#)] [[Google Scholar](#)]
9. Rifi L, Barkat A, El Khamlichi A, Boulaadas M, El Ouahabi A. Neurosurgical management of anterior meningo-encephaloceles about 60 cases. Pan Afr Med J. 2015;21:215. [[Article PMC gratuit](#)] [[PubMed](#)] [[Google Scholar](#)]
10. Sanoussi S, Chaibou M, Bawa M, Kelani A, Rabiou M. Encéphalocèle occipitale: aspects épidémiologiques, cliniques et thérapeutiques: à propos de 161 cas opérés en 9 ans à l'hôpital national de Niamey. Afr J Neurol Sci. 2009;28(1):24–9. [[Google Scholar](#)]
11. Hervey-Jumper SL, Cohen-Gadol AA, Maher CO. Neurosurgical management of congenital malformations of the brain. Neuroimaging Clin N Am. 2011;21(3):705–17. [[PubMed](#)] [[Google Scholar](#)]
12. Lo BW, Kulkarni AV, Rutka JT, Jea A, Drake JM, Lamberti-Pasculli M, et al. Clinical predictors of developmental outcome in patients with cephaloceles. J Neurosurg Pediatrics. 2008;2(4):254–7. [[PubMed](#)] [[Google Scholar](#)]

13. Bhandari S, Sayami JT, K C RR, Banjara MR. Prevalence of congenital defects including selected neural tube defects in Nepal: results from a health survey. BMC Pediatr. 2015;15:133. [[Article PMC gratuit](#)] [[PubMed](#)] [[Google Scholar](#)]
14. Sorri G, Mesfin E. Patterns of neural tube defects at two teaching hospitals in Addis Ababa, Ethiopia a three years retrospective study. Ethiop Med J. 2015 juill;53(3):119–26. [[PubMed](#)] [[Google Scholar](#)]
15. Radouani MA, Chahid N, Benmiloud L, Elammari L, Lahlou K, Barkat A. Epidémiologie et facteurs de risque des anomalies de fermeture du tube neural: données marocaines. Pan Afr Med J. 2015;22:43. [[Article PMC gratuit](#)] [[PubMed](#)] [[Google Scholar](#)]
16. Ndoma VN, Gaudeuille A, Nanguene J, Nghario JI, Issa-Mapoukaetude A. Des malformations du tube neural: spina bifida et encephalocele dans le service de chirurgie pédiatrique de Bangui. Rev CAMES Sci Santé 2016;4(1):13–6. [[Google Scholar](#)]
17. El Mhabrech H, Ben Mansour S, Dakkem M, Zrig A, Ben Hmida H, Hafsa C. Diagnostic anténatal de l'encéphalocèle. J Neuroradiol. 2016 mars;43(2):105–6. [[Google Scholar](#)]
18. Copp AJ, Stanier P, Greene NDE. Neural tube defects: recent advances, unsolved questions, and controverses. Lancet Neurol. 2013;12(8):799–810. [[Article PMC gratuit](#)] [[PubMed](#)] [[Google Scholar](#)]
19. Bergman JEH, Otten E, Verheij J, de Walle HEK. Folic acid supplementation influences the distribution of neural tube defect subtypes: a registry-based study. Reprod Toxicol. 2016;59:96–100. [[PubMed](#)] [[Google Scholar](#)]
20. Agha RA, Franchi T, Sohrabi C, Mathew G, for the SCARE Group. The SCARE 2020 Guideline: Updating Consensus Surgical Case Report (SCARE) Guidelines, International Journal of Surgery 2020;84:226-230.

