Clinical Presentations and Surgical Outcome In Patients With Occipital Encephalocele

Aurangzeb Kalhoro,[1*] Kashif Ahmed, [1] Abdul Sattar M Hashim [1]

ABSTRACT

Objective *To document the clinical presentations and outcomes of the surgical management in patients with occipital encephalocele.*

Study design *Descriptive case series.*

Place & Duration of study *Neurospinal and Cancer Care Institute Karachi, from June 2017 and October 2021.*

Methods *Patients with occipital encephalocele were included in this study. The demographic data and clinical presentations were recorded. The cognition function and developmental delays were assessed by the pediatric neurologist. Investigations performed included ultrasound, CT scan and MRI brain for the assessment of bony defect and the content of the encephalocele and brain. All patients were operated. At follow-up any associated complications and neurological development were recorded. The assessment of the outcomes included wound healing, head circumference size, symptoms of meningitis if any, and the presence of neurological deficit. Descriptive statistics were used to present the data.*

Results *A total of 21 patients with occipital encephalocele were managed during the study period. There were 12 male and 9 female patients. The age of the patients was between 4-days to 15-months with the mean age of 2.4-months. Enlarged head circumference with associated hydrocephalus was observed in four patients at presentation. One patient developed cerebrospinal fluid (CSF) leak. In one patient with Dandy Walker syndrome Y-connector was placed during surgery. The follow up continued for 18-months. A patient who developed CSF leak which did not stop spontaneously in postoperative period, the ventriculoperitoneal shunt was placed. No patient developed neurological deficits after surgery.*

Conclusion *Encephalocele is an infrequently encountered anomaly. Males were more often involved. Surgical procedure was tailored according to the content of the anomaly and associated conditions. CSF leak was observed in one patient who later developed hydrocephalus.*

Key words *Occipital encephalocele, Neural tube defect, CSF leak, Hydrocephalus, Neurological delay.*

INTRODUCTION:

Encephalocele, a congenital neural tube defect (NTD), involves the extrusion of intracranial matter, which may contain the brain, meninges, and, in some cases viable or non-viable tissue. This condition arises in the initial weeks of the fetal development due to the incomplete closure of the cranial portion of the neural tube. Its prevalence is reported to be 0.8 - 5.0 per 10,000 births.[1,2] The prognosis of the patient is based on the content of neural tissue and size of the sac of encephalocele.[3] Mortality occurs in occipital encephaloceles due to multiple associated anomalies of the brain and other organ systems. Those who survive, usually have seizures, cognitive deficits, spasticity and hydrocephalus. Counseling

[1] Neurosurgery Unit, Neurospinal Cancer Care Institute Karachi.

Correspondence:
Dr. Aurangzeb Kalhoro [1*]
Neurosurgery Unit
Neurospinal Cancer Care Institute
Karachi
Email: draurangzebkalhoro@gmail.com

of the family is important for all the issues that may be encountered.[4] The neurological outcome and IQ level are affected in addition to the associated chromosomal abnormalities and microcephaly.[5, 6]

There are many relevant investigations that are performed for this anomaly. It includes the ultrasound which is easily available. CT scan is required for the assessment of bony defects and MRI brain to document the content of the encephalocele sac. These are helpful in making the diagnosis as well as for planning the treatment.[7] Early diagnosis, management, and counseling play a crucial role in addressing this congenital brain malformations. Surgery, if necessary, can be performed as early as two to four months of age.[8,9] The morbidity and mortality of patients with encephalocele are quite variable but decreased in recent times due to the advancement in diagnostic tools, surgical techniques and postoperative care.[10] This study was conducted to document the clinical features and outcome of the surgical procedure performed for occipital encephalocele.

METHODS:

Study design, place & duration: A descriptive case series was conducted on patients with occipital encephalocele at Neurospinal Cancer Care Institute Karachi, from June 2017 to October 2021.

Ethical considerations: The consent was taken from the parents of the children. The study was approved by IRB of the institution. (No: 9721/18).

Inclusion criteria: All patients with occipital swelling that was noted at birth with the herniation of intracranial content secondary to a primary osseous defect, diagnosed with CT scan and magnetic resonance imaging of the brain, were included.

Exclusion criteria: Patients who had previous surgery for the anomaly, those with ruptured sac, and a sac containing a large amount of cerebral tissue were excluded from the study. Patients with other associated anomalies were also not enrolled.

Sample size estimation and sampling technique: All patients managed during the study period were included with non-probability consecutive sampling technique.

Study protocol: The demographic and clinical data of all the study patients were collected on a pre designed form. It included the age, gender, anatomical location and size of the encephalocele, the known comorbid such as episodes of fits or seizure, and hydrocephalus. The cognition function and developmental delays were assessed by the pediatric neurologist. The surgical procedure performed and postoperative complications were also noted. Follow up visits were encouraged to document postoperative progress.

Surgical technique:
The patient was kept in a prone position so as to provide optimal access to the occipital region while ensuring proper monitoring of the vital signs. A careful incision was made over the occipital region to expose the encephalocele sac and the surrounding tissues. The dura mater was opened to expose the brain and the encephalocele. The neural tissue within the encephalocele was carefully examined for any abnormalities. The redundant neural tissue was resected. If there was any tethering of the spinal cord or nerves, it was addressed during the surgery. The dura mater was then closed using sutures to provide a protective barrier for the brain. The skull defect was repaired using various techniques, such as bone grafts or synthetic materials. The incision was closed in layers. After surgery, close monitoring was done in the intensive care unit. Antibiotics were administered to prevent infection, and pain management was provided.

Statistical analysis: Descriptive statistics were used to present data in number and percentages.

RESULTS:

A total of 21 patients were managed. It included twelve males and nine females. The mean age of the patients was 2.4-months (from 4-days to 15-months). Five patients had an enlarged head circumference with associated hydrocephalus, of which four had preoperative hydrocephalus for which ventriculoperitoneal shunts were placed. One patient developed hydrocephalus after encephalocele surgery (10^{th} post-surgery day). A patient had Dandy-Walker cyst that was treated with a Y-connector. Additionally, one patient had strabismus since birth, and developmental delay was suspected in one patient.

Seizures were reported in four patients. These were successfully treated with a single epileptic medication. The sac size varied from 3.4 cm × 2.5 cm to 20 cm × 10 cm. History of consanguinity was observed in six patients. No patient developed neurological deficits after surgery. There were no instances of loss of vision, spastic paralysis, or ataxia. One patient presented with cerebrospinal fluid (CSF) leak within two weeks of surgery, and the same patient developed

Table I: Characteristics of the Patients (n=21)

Characteristics	n	%
Gender		
Male	12	57.1%
Female	09	42.9%
Hydrocephalus		
Pre surgery	04	19.0%
Post surgery	01	4.76%
Dandy Walker Syndrome	01	4.76%
Cousin marriage	01	4.76%
Postoperative Complication		
CSF leak	01	4.76%

postoperative hydrocephalus. After CSF culture, a ventriculoperitoneal shunt was successfully placed (table I).

DISCUSSION:

Encephalocele is an uncommon anomaly which is also revealed in this study. The average size of the sac was from 3.5 cm × 2.5 cm to 20 cm × 10 cm in diameter. The sac content mainly consisted of dysplastic diverticulum, while some contained brain tissue in the degenerated form. The bony defect was proportional to the size of the sac, with some patients having a smaller base and a larger sac. No CSF leakage was observed except in one patient after surgery that were typically performed after two months of age. This specific timing was chosen to ensure the health of the infants. The anesthesia team protocol was followed to minimize complications relevant to the infantile age group. During surgery, viable brain tissue and blood vessels were preserved and carefully pushed under the dura.

In a study, males were predominantly affected, contrasting with some studies indicating a female predominance.[11,12] Males were more frequently managed in our study as well. In a study on occipital encephalocele it was demonstrated that the safe removal of dysplastic brain tissue with the preservation of vessels without the need of advance procedures was possible. Our approach mirrored this plan, focusing on the removal of dysplastic nonviable tissue and vasculature preservation. Proper dural closure was emphasized to minimize cerebrospinal fluid leaks.[13]

The groundbreaking realm of intrauterine repair for encephalocele signifies a major step in mitigating the sac herniation progression, offering a glimpse into the potential for reversing microcephaly. Examining the efficacy of such procedures necessitates a meticulous examination of outcomes, considering immediate surgical success and the long-term neurodevelopmental trajectory of affected individuals.[14,15] Although intrauterine repair is performed in a specialized center it is hoped that this approach may be widely available in future. Antenatal ultrasounds are not performed routinely in Pakistan in many geographical regions. They are usually done for obstetrics reasons and late in the pregnancy. Thus intrauterine repair may have limited application in our region.

Occipital encephalocele is often associated with hydrocephalus, potentially leading to a worse clinical outcome. Families require counselling based on various aspects and the outcome.[16,17] In another study, 64.7% of cases had hydrocephalus associated with occipital encephalocele, with 36.4% diagnosed before surgery and 63.6% developing hydrocephalus postoperatively.[6] In our study, five patients had hydrocephalus, with ventriculoperitoneal shunt placement preoperatively in four cases. One patient developed hydrocephalus post-surgery, requiring a ventriculoperitoneal shunt, and no postoperative infections were reported in our series.

Studies have reported that patients with occipital encephalocele require surgery. A potential link is noted between TORCH infections and the anomaly.[18,19] Consanguinity may also contribute to encephalocele development, requiring further investigations through larger studies. In our study, at follow-up, no mortality was reported. Seizures were observed in four patients for which appropriate antiepileptic medications were used. A study demonstrated good seizure control with post-surgery anti-seizure medications, with a mortality rate of 5.9%.[20] This differ from our study due to the large number of patients**.** Results of encephalocele surgery show improve trends in a study from Pakistan with reduction in mortality and morbidity.[21]

Limitations of the study: The encephalocele is a rare condition. The number of patients in our study are therefore limited.

CONCLUSION:

Encephalocele poses a complex neurosurgical challenge. Various issues associated with this condition demand careful management that are tailored for each patient, before as well as after surgery, like hydrocephalus.

REFERENCES:

1. Ghritlaharey RK. A brief review of giant occipital encephaloceles. J Neurosci Rural Pract. 2018;9(4):455. DOI: 10.4103/jnrp.jnrp_189_18

2. Kýymaz N, Yýlmaz N, Demir I, Keskin S. Prognostic factors in patients with occipital encephalocele. Pediatr Neurosurg. 2010;46:6-11. https://doi.org/10.1159/000314051

3. Bozinov O, Tirakotai W, Sure U, Bertalanffy H. Surgical closure and reconstruction of a large occipital encephalocele without parenchymal excision. Childs Nerv Syst. 2005;21:144-7. doi: 10.1007/s00381-004-1020-5.

4. Gandhoke GS, Goldschmidt E, Kellogg R, Greene S. Encephalocele development from a congenital meningocele: case report. J Neurosurg Pediatr. 2017;20:419-22. doi: 10.3171/2017.6.PEDS17178.

5. Simair IA, Ali J, Khattak A, Qureshi A, Salah-ud-Din T. Outcome comparison of endoscopic (endonasal trans-sphenoidal) repairs of CSS leak vs. transcranial approach. Pakistan J Neurol Surg. 2021;25:18-28. DOI: https://doi.org/10.36552/pjns.v25i1.527

6. Nagy MR, Saleh AE. Hydrocephalus associated with occipital encephalocele: surgical management and clinical outcome. Egypt J Neurosurg. 2021;36:1-7. https://doi.org/10.1186/s41984-021-00101-5

7. Naidich TP, Altman NR, Braffman BH, McLone DG, Zimmerman RA. Cephaloceles and related malformations. Am J Neuroradiol. 1992;13:655-90.

8. Cruz AJ, De Jesus O. Encephalocele. StatPearls Publishing. [Internet] Available from URL https://www.ncbi.nlm.nih.gov/books/NBK562168/ accessed on September 2023

9. Markovic I, Bosnjakovic P, Milenkovic Z. Occipital encephalocele: Cause, incidence, neuroimaging and surgical management. Curr Pediatr Rev. 2020;16:200-5. Êhttps://doi.org/10.2174/1573396315666191018161535

10. Bui CJ, Tubbs RS, Shannon CN, Acakpo-Satchivi L, Wellons JC, Blount JP, et al. Institutional experience with cranial vault encephaloceles. J Neurosurg Pediatr. 20071;107:22-5. https://doi.org/10.3171/PED-07/07/022

11. Wen S, Ethen M, Langlois PH, Mitchell LE. Prevalence of encephalocele in Texas, 1999–2002. Am J Med Genet A. 2007;143:2150-5. https://doi.org/10.1002/ajmg.a.31907

12. Nath HD, Mahapatra AK, Gunawat P. A torcular encephalocele with proatlas defect and os-terminale. Asian J Neurosurg. 2012;7:84-6. DOI: 10.4103/1793-5482.98652

13. Alqadhi S, Khan UA. Surgical management and clinical outcome of occipital encephalocoele. J Uni Med Dent Coll. 2017;8:6-10.

14. Gupta N. Fetal repair of encephaloceles. Journal of Neurosurg Pediatr. 2020;26:603-4.https://doi.org/10.3171/ 2020.5.PEDS20234

15. Timur H, Sanhal CY, Tokmak A, Müftüoglu KH, Danisman N. Prenatal diagnosis of tectocerebellar dysraphia with occipital encephalocele. J Clin Diag Res. 2015;9(12):QD05. doi:10.7860/JCDR/2015/16128.6987

16. Refaee EAE, Refaat MI, Reda M. Incidence of secondary hydrocephalus after excision of huge encephaloceles in neonates: case study. J Neurol Surg A Cent Eur Neurosurg. 2018;79:15-8. doi: 10.1055/s-0036-1597548.

17. Zabsonre DS, Lankoande H, Zoungrana-Ouattara CF, Thiombiano A, Ouedraogo S, Sanou A, et al. association of hydrocephalus with neural tube defect: our experience with the surgical treatment in one or in two operative stages (on separate days). Pediatr Neurosurg. 2019;54:1-5. https://doi.org/10.1159/000494562

18. Ramos-Fresnedo A, Domingo RA, McGeary RC, Sirven JI, Feyissa AM, Tatum W, et al. Encephalocele-associated drug resistant epilepsy of adult-onset: diagnosis, management, and outcomes. World Neurosurg. 2021;151:91-101. https://doi.org/10.1016/j.wneu.2021.04.121

19. Yucetas SC, Uçler N. A retrospective analysis of neonatal encephalocele predisposing factors and outcomes. Pediatr Neurosurg. 2017;52:73-6. https://doi.org/10.1159/000452805

20. Da Silva SL, Jeelani Y, Dang H, Krieger MD, McComb JG. Risk factors for hydrocephalus and neurological deficit in children born with an encephalocele. J Neurosurg Pediatr. 2015 ;15:392-8. https://doi.org/10.3171/2014.10.PEDS14192

21. Rehman L, Farooq G, Bukhari I. Neurosurgical interventions for occipital encephalocele. Asian J Neurosurg. 2018;13:233-7. DOI: 10.4103/1793-5482.228549

Received for publication: 10-12-2023
Sent for revision: 23-01-2024
Accepted after revision: 27-01-2024

Author's contributions:
Aurangzeb Kalhoro: Conception, study design, data collection, statistical analysis and manuscript writing and revising. Kashif Ahmed: Data analysis, data interpretation and literature review.
Abdul Sattar M Hashim: Study design, data analysis and interpretation.

All authors agreed to be accountable for the content of the article.

How to cite this article:
Kalhoro A, Ahmed K, Hashim ASM. Clinical presentations and surgical outcome in patients with occipital encephalocele J Surg Pakistan. 2023;28 (4):100-4.

Ethics statement: Institutional review board permission was obtained prior to the study and informed consent were taken from the parents.

Competing interest: Authors declare that they have no competing interests.

Source of funding: None

Disclosure: Nil