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Clinico-radiological evaluation, demographic pattern and surgical management in patients of occipital encephalocele.

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Author's contribution :
MJ- Definition of intellectual content, Literature survey, Prepared first draft of manuscript, Implementation of study protocol, data collection, data analysis, manuscript preparation and submission of article, **KVK-** Concept, design , clinical protocol, manuscript preparation, editing and revision, statistical analysis, **SA[3]-** Review manuscript, preparation of tables, **SA[4-]** Manuscript revision **KA-** Coordination and manuscript revision

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Patients who lost follow up after one month are not included for comparing the outcome in the study.

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Abstract

Background -
Encephalocele is a birth defect caused by a neural tube defect in which a portion of the brain protrudes through an opening in the skull. This can cause a swelling or sac like protrusion over the affected area. Occipital encephalocele is the most common type and typically appears as a midline swelling over the back of the skull. It is important to diagnose and treat this condition as early as possible to prevent complications and improve outcomes. Treatment may involve surgery to repair the opening in the skull and reposition the protruding brain tissue.

AIMS AND OBJECTIVES - The aim of this review is to provide a comprehensive overview of the current understanding of occipital encephalocele, including its incidence, signs and symptoms, diagnosis, and treatment. By reviewing the existing literature this paper aims to present a consolidated and up-to-date understanding of this congenital condition.

Material and methods - This retrospective study included 24 patients admitted with congenital swelling over occipital scalp under department of neurosurgery , GRMC and associated J.A. group of Hospitals from May 2022 to April 2023 who underwent surgery. No randomisation done.

Results : The study included 24 patients, with 71% being female and 29% being male. Occipital encephalocele has an incidence rate of 1 in 3000 to 1 in 10,000 live births, and about 90% of cases involve the midline. Magnetic resonance imaging (MRI) is considered the best diagnostic method for occipital encephalocele. Surgical intervention is the preferred treatment option for occipital encephalocele. However, the overall morbidity and mortality rate associated with this condition is still high, despite advanced surgical techniques. Nevertheless, recent advancements in high-resolution imaging, proper surgical management, and post-operative care have significantly improved outcomes.

CONCLUSION- Occipital encephalocele is the most common type of encephalocele. Diagnosis primarily relies on neuroimaging. Surgery is the optimal treatment. Despite high morbidity and mortality rates, advancements in imaging, surgical techniques, and post-operative care have led to significant improvements in recent years.

Key words- Encephalocele, Occipital encephalocele, Neural tube defect, meningoencephaloceles, Posterior encephaloceles

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INTRODUCTION:

Encephalocele is a congenital defect characterised by the extracranial protrusion of intracranial structures through a skull opening. It results from the incomplete separation of surface ectoderm and neuroectoderm during embryonic development. The protruded brain and meninges are typically covered by a skin or thin membrane, resembling a sac-like structure. Encephalocele defects are classified into four types based on the content protruding through the skull opening: meningoencephaloceles (herniations of cerebrospinal fluid, brain tissue and meninges), meningoceles (herniations of the meninges and CSF), glioceles (glial-lined cyst containing CSF), and atretic cephaloceles (comprising of dura, fibrous tissue and degenerated brain tissue). Patients with encephalocele have a high risk (60-80%) of associated structural abnormalities and a 60% chance of developing hydrocephalus (1).

Various chromosomal abnormalities, including monosomy X and trisomies such as 13, 18, and 21, as well as over 30 different syndromes, can be associated with encephalocele [2]. Occipital encephalocele is characterised by a midline swelling over the occipital bone, more commonly seen in females [3]. It can range from a small opening to a giant sac larger than the newborn's head [4]. Encephalocele is often accompanied by other congenital anomalies or genetic syndromes, with 60% of cases having additional malformations or chromosomal defects. The two types of occipital encephalocele are supratorcular and infratorcular, with protrusion of different intracranial structures, such as meninges, occipital lobes, or ventricles [5]. The skull defect can extend to the posterior lip of the foramen magnum and the posterior arch of the atlas. Almost 15%-20% of newborns will have other anomalies, including neural tube defects, microcephaly, Arnold- Chiari malformations Type 2 or 3, craniosynostosis, and syringomyelia [6].

Poor prognostic features include large size of the sac, significant brain herniation and abnormality of underlying brain. Hydrocephaly and ventriculomegaly with hydrocephalus, and infection are common complications encountered in the post operative period.

The exact cause of encephalocele is not fully understood, but it is likely influenced by a combination of genetic and environmental factors. Family history of neural tube defects increases the risk. The CEP290 gene has been identified as being associated with occipital encephalocele. Environmental factors such as hyperthermia, aflatoxins, maternal malnutrition, and certain toxins or infections may also contribute to its development [7]. The role of maternal folate levels in encephalocele is not clear, although adequate intake of folic acid during pregnancy (400 micrograms of folic acid daily) is recommended to reduce the risk of neural tube defects [8].

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Encephalocele account for 10% - 20% of all craniospinal dysraphism [4]. The published estimates vary in the range of 1/5000 live births to approximately 1 in 10,000 babies each year; or about 0.8–5.6/10,000 live births [1, 6, 8, 9]. Occipital encephalocele is the most common form of the anomaly and is present in the range of 80-90% of all encephalocele. In the USA, the incidence is between 1 in 3000 to 1 in 10,000 live births; approximately 90% of them involve the midline [1, 10], and 70% of them occur in females [4].

The signs and symptoms of occipital encephalocele can vary widely based on factors such as the size and location of the defect, the amount and type of brain tissue protruding through the defect, and associated conditions affecting cerebrospinal fluid circulation or other anomalies commonly seen in individuals with this congenital disorder. The modified classification as proposed by Rosen Field as shown in table 1 was used to classify the encephalocele anatomically.

Table 1. Encephalocele classification :

Convexity	Occipital Parietal Sagital Occipitocervical
Sincipital	Frontoethmoidal Nasofrontal Nasoethmoidal Nasoorbiral Inter frontal Craniofacial cleft
Basal	Intra nasal Sphenoorbital Sphenomaxillary Sphenopharyngeal
Atretic	

Material and methods:

This was a retrospective study on 24 patients who were admitted with congenital swelling over occipital scalp under department of neurosurgery, GRMC and associated J.A. group of Hospitals, Gwalior from May 2022 to April 2023 who

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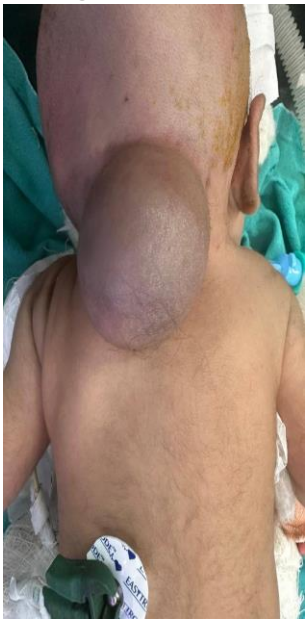
underwent surgery and after analysing the patient's outcome the results were obtained.

After the admission of patients, they underwent clinical (Figure 1) and radiological examination and diagnosis of occipital encephalocele relies on the use of ultrasound, computed tomography (CT) (Figure 2), and magnetic resonance imaging (MRI)(Figure 3**)** with MR venography and MR angiography [11]. Pre-operatively all the patients were investigated thoroughly following which after assessment of all the patients they underwent surgical procedure. Intra-operative findings, surgical procedures and operative complications were noted. Demographic, clinical, radiological and operative data were reviewed from hospital charts.

The following data points were collected: patient's age and sex, location and size of encephalocele, size of skull defect, presence of associated congenital anomalies, type of surgical procedure performed, whether skull defect repair was conducted, occurrence and size of any recurrence, cosmetic acceptability, surgical complications during and after the operation, and outcomes during the follow-up period. Detailed neurological examination was performed noting specifically presence or absence of any neurological deficit.



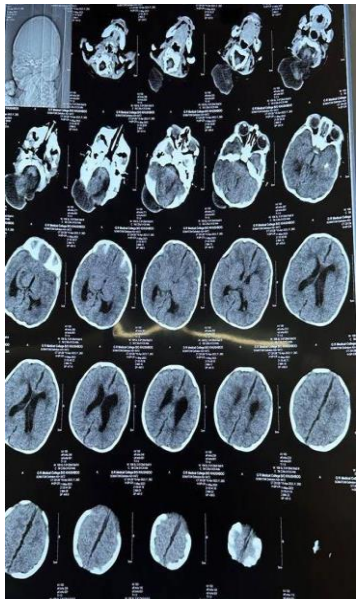


Figure 1 **Figure 2**

Figure 1 and 2 - Case of occipital encephalocele and pre op CT scan

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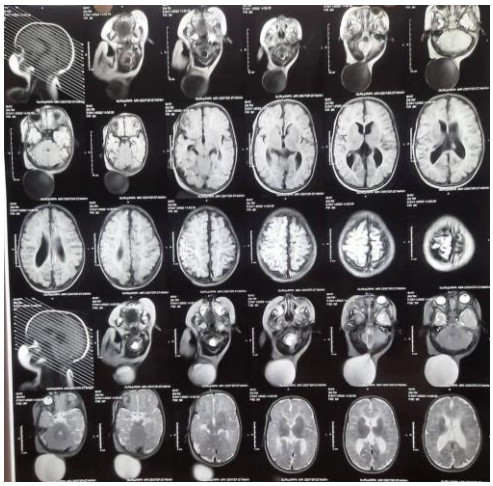

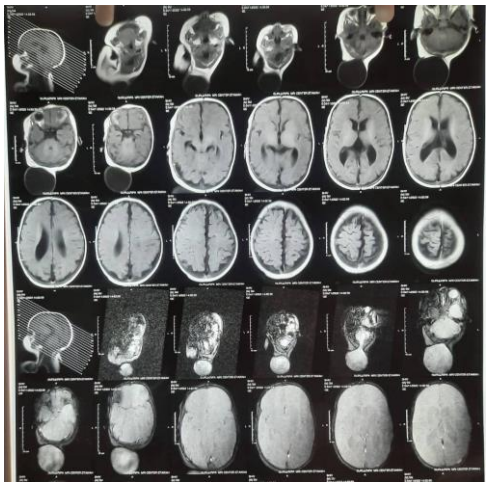

Figure 3

Figure 3 - Pre op MRI scan

Inclusion criteria :
All the patients with clinical and radiological diagnosis of occipital encephalocele and planned for surgery.

Exclusion criteria :
Patients who were born before the defined period of study and those not willing for participation.

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RESULTS :

A non-randomised, retrospective, single institute study was conducted between May 2022 to April 2023, in GR Medical College and JA group of hospitals (GRMC), Gwalior over a period of one year in the department of Neurosurgery on 24 patients diagnosed with occipital encephalocele. Based on the data collected and data analyses following observations were made.

In this study, 24 patients were included, comprising 17 females and 7 males. The average age at the time of presentation was 10 months. The surgical approach for encephalocele varied based on its location and type (Table: 2). Direct extracranial repair was feasible for frontonasal, cranial vault, and occipital encephaloceles, while intracranial repair may be necessary for nasoethmoidal and nasoorbital types. For older children with larger encephaloceles, a collaborative effort between a neurosurgeon and a plastic surgeon was required for effective treatment.

Table: 2 - Surgical approach for encephalocele

Surgical Approach	No. of Cases	Percentage
Direct excision and repair	18	75
Craniotomy and repair	1	4.1
Associated Hydrocephalus Requiring VP shunt	5	20.9
Total	24	100

DISCUSSION:

Encephalocele is a type of craniospinal dysraphism characterised by a protrusion of brain tissue through a defect in the skull. It accounts for approximately 10 to 20% of all craniospinal dysraphism cases. Occipital encephalocele, specifically, refers to encephalocele occurring at the back of the skull, and it is more commonly seen in females, comprising about 70% of cases while in our study similar results were obtained with the female incidence of 70.8%. (12).

The lesions associated with encephalocele can be covered by normal skin, dysplastic skin (abnormal skin development), or a thin and distorted meningeal membrane (the protective membrane surrounding the brain). The size of the encephalocele can vary,

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and larger swellings may involve significant brain herniation, abnormalities in the underlying brain structure, microcephaly (abnormally small head), or ventriculomegaly (enlarged brain ventricles) (12).

Treatment of encephalocele depends on the size and characteristics of the lesion. Encephaloceles with a small amount of dysfunctional brain tissue can often be managed by surgically removing the herniated brain tissue and repairing the dural defect (the defect in the protective covering of the brain). However, in cases where there is a large defect with a considerable amount of herniated brain matter, surgical management becomes more challenging (12).

In such situations, the preservation of the herniated brain tissue may be attempted, and an expansible cranioplasty can be performed. Expansible cranioplasty involves the use of materials or techniques that allow for the gradual expansion of the skull over time, accommodating the herniated brain tissue. The primary goal of surgery is to achieve a watertight closure of the dural defect after reducing the herniated brain tissue. Additionally, the surgery aims to address the aesthetic and functional deformities resulting from the encephalocele and its treatment (12).

Amongst newborns with occipital encephalocele seen in studies of Kanesen D et al (6), Rehman L et al (8) and Satyarthee GD et al (10) 60-70 % of the patients are at risk of developing hydrocephalus, which should be treated by application of the ventriculoperitoneal (VP) shunt. In the majority of cases, hydrocephalus develops post-operatively. One of the mechanisms causing hydrocephalus may be torsion of the aqueduct of Sylvius or aqueductal stenosis. Surgical repair of the occipital encephalocele sac may provoke hydrocephalus due to changes in the CSF dynamics (10).

CONCLUSION :

Early diagnosis and treatment of encephalocele can significantly improve outcomes. Different surgical approaches may be necessary depending on the location and type of the encephalocele. For certain types, such as occipital, parietal, frontal, and frontonasal encephaloceles, direct extracranial repair may be possible without opening the cranium. However, for sincipital and basal encephaloceles, craniotomy is usually necessary. The successful management of encephalocele requires careful consideration of technical aspects of the surgical approach, as well as coordination between neurosurgeons and plastic surgeons, particularly in cases involving larger encephaloceles in older children.

References:

1. Ugras M, Kavak O, Alpay F, Karabekir SH, Bicer S. New Born Children with Encephalocele Journal of Neurology and Neuroscience 2016; 7(73): 1-4.

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2. Franco A, Jo SY, Mehta AS, Pandya DJ, Yang CW. A Rare Triad of Giant Occipital Encephalocele with Lipomyelomeningocele, Tetralogy of Fallot, and Situs Inversus. J Radiol Case Rep 2016; 10(3): 36-46

3. Verma SK, Satyarthee GD, Singh PK. Pediatr Neurosci 2013; 8(3): 207-9. http://dx.doi.org/10.4103/1817-1745.123666.

4. Agarwal A, Chandak A V , Kakani A, Reddy S. A giant occipital encephalocele. APSP J Case Rep 2010; 1(2): 16. PMID: 22953259

5. Nath HD, Mahapatra AK, Borkar SA. A giant occipital encephalocele with spontaneous hemorrhage into the sac: A rare case report. Asian J Neurosurg 2014; 9(3): 158-60. http://dx.doi.org/10.4103/1793-5482.142736 PMID: 25685207

6. Kanesen D, Rosman AK, Kandasamy R, Giant occipital encephalocele with chiari malformation type 3. J Neurosci Rural Pract 2018; 9(4): 619-21.

7. Agthong S, Wiwanitkit V. Encephalomeningocele cases over 10 years in Thailand: a case series. BMC Neurol 2002; 2: 3. http://dx.doi.org/10.1186/1471-2377-2-3 PMID: 12010577

8. Rehman L, Farooq G, Bukhari I. Neurosurgical Interventions for Occipital Encephalocele. Asian J Neurosurg 2018; 13(2): 233-7. http://dx.doi.org/10.4103/1793-5482.228549 PMID: 29682014

9. Kumar V, Kulwant SB, Saurabh S, Richa SC. Giant occipital meningoencephalocele in a neonate: A therapeutic challenge. J Pediatr Neurosci 2017; 12(1): 46-8. http://dx.doi.org/10.4103/1817-1745.205655 PMID: 28553380

10. Satyarthee GD, Moscote-Salazar LR, Escobar-Hernandez N, et al. A giant occipital encephalocele in neonate with spontaneous hemorrhage into the encephalocele sac: Surgical management. J Pe- diatr Neurosci 2017; 12(3): 268-70.

11. Tortori-Donati P, Rossi A, Biancheri R. Pediatric Neuroradiology, Brain. In: Chapter 4, Syndromes with Cephaloceles. Springer- Verlag: Heidelberg Berlin 2005; p. 80.

12. MUHAMMAD AFZAL SHEIKH, MUHAMMAD IBRAHIM, AHMED NADEEM TAHIR. Occipital Encephalocele and Review of Literature. Pak. J. of Neurol. Surg. – Vol. 18, No. 1, Jan. – Jun., 2014