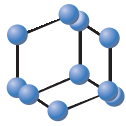


REVIEW ARTICLE


**BENTHAM
SCIENCE**

Occipital Encephalocele: Cause, Incidence, Neuroimaging and Surgical Management


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Abstract: Aims: To review and present the current knowledge of incidence, signs and symptoms, diagnosis and treatment of the occipital encephalocele.

Background: Encephalocele (E) is a defect of the neural tube that refers to congenital malformations featured by skull defect and dura with extracranial spread of intracranial structures. Occipital encephalocele (OE) are the most common form of this congenital disorder and are manifested as a swelling of different sizes over the occipital bone in the midline. Proper diagnosis and treatment is highly important in the management of this congenital malformation of brain.

Objective: To review and present the current knowledge of incidence, signs and symptoms, diagnosis and treatment of the occipital encephalocele

Methods: We conducted a search of case reports or case-series of patients by the use of electronic databases: Pub Med, Medline, Index Medicus, Scopus. The key words were: encephalocele, occipital encephalocele, neural tube defect, congenital malformation. The search was updated to December 31, 2018. Papers published in English were the only source of information.

Results: Occipital encephalocele are more frequent in females than in males. The incidence is between 1 in 3000 to 1 in 10,000 live births; approximately 90% of them involve the midline. Magnetic resonance imaging is the method of choice in diagnosis and surgery is the best option for the treatment of OE. Overall morbidity and mortality is still high in spite of advanced surgical management, but have been significantly improved in recent years thanks to sophisticated high-resolution imaging, adequate and proper surgical treatment and decent post-operative care.

Conclusion: Occipital encephalocele is the most common form of encephalocele. The diagnosis is mostly based by the use of neuroimaging techniques. Operation is the best option for treatment. Overall morbidity and mortality is still high, but have been significantly improved in recent years thanks to sophisticated high-resolution imaging, adequate and proper surgical treatment and decent post-operative care.

Keywords: Occipital encephalocele, cause, incidence, neuroimaging, cerebrospinal fluid, neuroimaging techniques.

1. INTRODUCTION

Encephalocele (E) is a defect of the neural tube that refers to congenital malformations characterized by skull opening and dura with an extracranial spread of intracranial structures. It is an embryonic mesodermal anomaly that results in the failure of the separation of the surface ectoderm from the neuroectoderm. The disorder is manifested by the protrusion of the brain with or without the protrusion of the meninges through an existing defect in the skull. A part of the brain that is held outside the skull is usually covered with

a skin or thin membrane so that the defect resembles a small sac. Depending on the intracranial content which protrudes through the skull opening, E defects are divided into four types: meningoencephaloceles (herniations of cerebrospinal fluid (CSF), 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 60-80% risk of structural abnormalities, and 60% will develop hydrocephalus [1]. Many chromosomal abnormalities are associated with neural tube defects, such as monosomy X; numerous trisomies including 13, 18, and 21, as well as lesser-known 5, 7, 8, 11 mosaicism, 14, 15, 16, and 20 mosaicism, and tetraploidy [2]. Encephalocele is also a part of more than 30 different

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syndromes including Meckel syndrome, Fraser syndrome, Roberts syndrome, and Walker-Warburg syndrome, Amniotic band syndrome, Knobloch syndrome [3].

Occipital encephalocele (OE) is manifested as swelling of different sizes over the occipital bone in the midline, and is more frequent in females than in males [4] (Fig. 1). Usually, the head size is small [5]. Bony defect of the skull can vary in size from a few millimeters to more than 20 centimeters; sometimes, the sac is bigger than the newborn's head and is labeled as a giant OE [6] (Fig. 2). In one study, 16% of children harbored OE longer than 20 cm [7]. Although the lesion may be presented as an isolated disorder, it is often associated with additional congenital anomalies or genetic syndromes [2, 8]. In 60% of children with OE, another malformation and/or chromosomal defect was diagnosed [9, 10] and almost 15%-20% of newborns will have other anomalies, including neural tube defects, microcephaly, Arnold-Chiari malformations Type 2 or 3, craniostyostosis, and syringomyelia [11] (Fig. 3). Occipital encephalocele, typically localised in the midline of the occipital bone between the lambda and foramen magnum, exhibits two types: the supratentorial and the infratentorial one. The intracranial structures protruded in encephalocele's sac may be

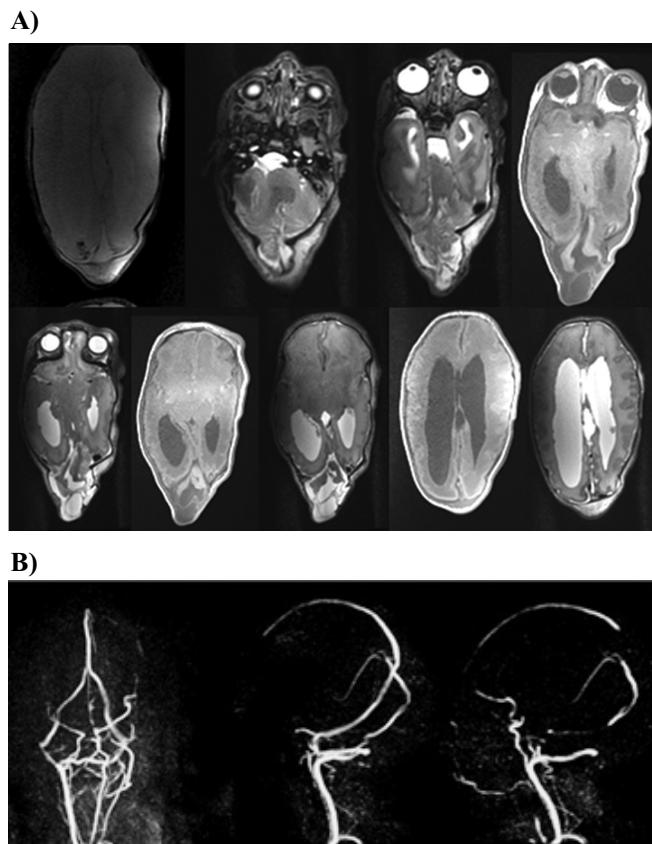


Fig. (1). Occipital encephalocele with distortion of brain stem and cerebellum. **A:** Axial T1W and T2W including T2* images show occipital cephalocele presented with big calvarial defect and stretched distorted brain stem and cerebellum with vermis; lateral ventricles are dilated with bilaterally present subependymal micro-nodular heterotopia of grey matter. **B-** 2D TOF MR Venogram shows elevated straight sinus which corresponds in location to a falcine sinus. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

different including meninges, occipital lobes, or ventricles, infrequently cerebellum, brainstem, or torcula [5]. The occipital bone defect sometimes can extend its opening to the posterior lip of the foramen magnum and even up to the posterior arch of the atlas [4].

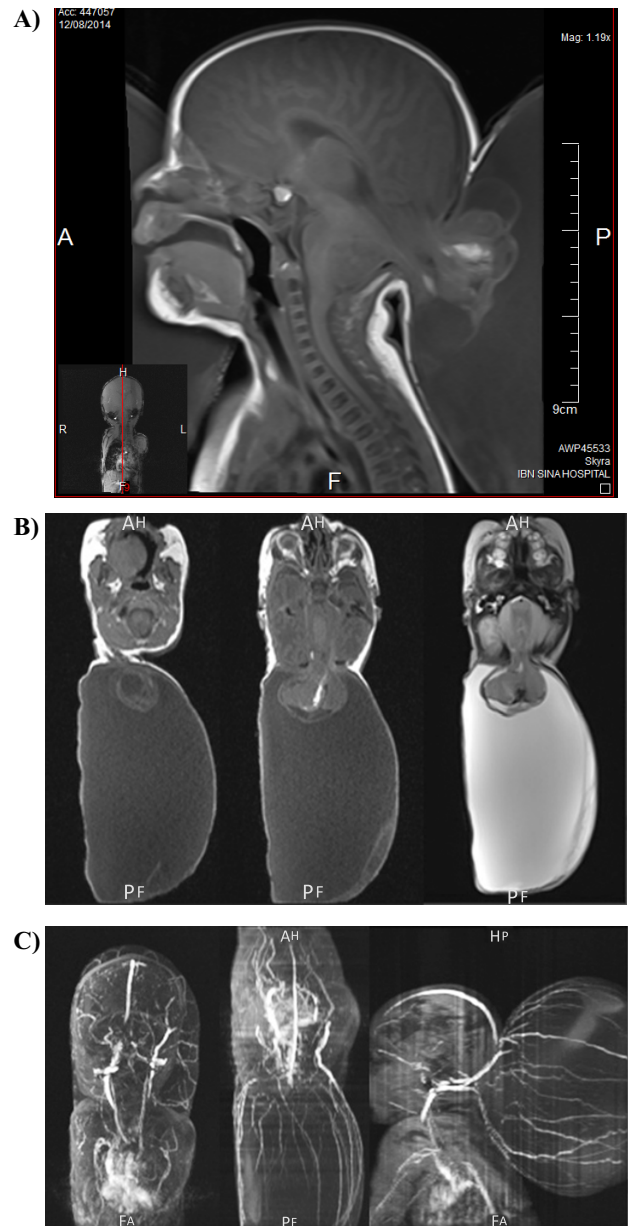


Fig. (2). Giant occipital encephalocele. **A)** Sagittal T1W image shows a large, giant occipital cephalocele with supratentorial and infratentorial brain tissue which enter cephalocele sac along with CSF from corresponding cisterns and forth ventricle; T1W hyperintensity within distorted and stretched brain tissue correlates with hemorrhage (blood) versus slow blood flow in sinuses. **B)** Axial T1W and T2W images show supratentorial and infratentorial brain tissue stretched posteriorly within mostly cystic formation extends through occipital bone defect; different, slow blood flow visible as T1W hyperintense linear structure due to stretched, elongated venous sinuses. **C)** Two axial and one sagittal views of 2D TOF venogram show dural venous sinuses and pathologic venous drainage into cephalocele sac. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

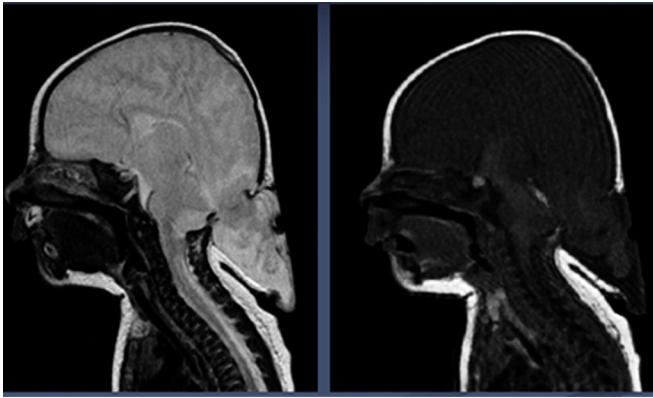


Fig. (3). Chiari III malformation, midsagittal T2W and T1W images present displaced and stretched brain tissue (cerebellum and brain stem) and corresponding CSF spaces which are protruded (herniated) through dorsal calvarial (osseous) defect in lower occipital bone with preserved alignment and continuity of upper cervical column in which case might be considered as occipital cephalocele rather than Chiari III malformation; T1W hyperintensity in posterior fossa as hemorrhagic content in part of distorted CSF spaces. (A higher resolution / colour version of this figure is available in the electronic copy of the article).

2. CAUSE OF THE MALFORMATION

The exact cause of E is unknown, and it is probably multifactorial, including both genetic and environmental factors. Individuals with a family history of neural tube defects (spina bifida, anencephaly) are more likely to develop encephalocele. Hypothetically, individuals with the genetic predisposition of neural tube defects might have the same predisposition to the development of E. Presence of the gene associated with certain disorders does not mean the development of the anomaly. However, the 'activation' of some factors (e.g. environmental) might trigger the disease [3]. An important gene associated with occipital encephalocele is CEP290 (Centrosomal Protein 290) [12]. Certain researchers have emphasized that non-specific environmental factors (such as hyperthermia, aflatoxins, genetic background, mother's lack of nutrition or other environmental factors) are involved in the development of E [13-16]. Even certain toxins or infections might play a role in its evolution [3]. Maternal levels of folate, as a protective factor, are relevant to myelomeningocele but its relevance for E is still 'not clear' [17]. The use of folic acid (400 micrograms of folic acid daily) in the diet of the pregnant women 'might lower the risk of some neural tube defect' [3].

3. INCIDENCE

Encephalocele account for 10% - 20% of all craniospinal dysraphism [6]. 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, 3, 11, 17, 18]. Occipital encephalocele is the most common form of the anomaly and is present in the range of 80-90% of all (E). In the USA, the incidence is between 1 in 3000 to 1 in 10,000 live births; approximately 90% of them involve the midline [1-3, 19], and 70% of them occur in females [6, 7]. Solitary encephalocele is a sporadic phenomenon and many of them

may be part of the genetic syndrome (Meckel-Gruber syndrome, Von Voss-Cherries syndrome, Valker-Varburg syndrome, Knobloch syndrome, Joubert syndrome, Valker Varburg syndrome) [2, 6, 19]. Hydrocephalus is an important clinical manifestation and is estimated to be identified in about 60% of all newborns with OE [4].

4. SIGNS AND SYMPTOMS

Signs and symptoms of OE may vary significantly depending on its size and localization, as well as the amount and type of brain tissue that protrudes through the bone defect, conditions related to the circulation of CSF and/or other anomalies which are very common in individuals with this congenital disorder. Posterior encephaloceles are frequently associated with neurological problems. Some infants may be asymptomatic on physical examination [6, 11, 19], but others might harbor a numerous and different signs and symptoms such as delays in reaching developmental milestones, intellectual disability, learning disabilities, growth delays, seizures, vision impairment, uncoordinated voluntary movements (ataxia), hydrocephalus, spastic paraplegia or quadriplegia and microcephaly [3, 5, 17].

5. DIAGNOSIS

The diagnosis of OE relies on the use of ultrasound, Computed Tomography (CT), and Magnetic Resonance Imaging (MRI) with MR venography and MR angiography [20].

6. ULTRASONOGRAPHY

The ultrasonography has been applied extensively in the prenatal diagnosis of OE. Its efficacy is based on the capacity to identify and visualizes a cranial defect and 'the varying degree of brain herniation,' but the mother's body habitus, the position of the fetus and the surrounding amniotic fluid have been found to be the hindering factors in the fine analysis of the brain and /or CNS. The skull defect can be demonstrated in 80% of fetuses harboring OE [21].

7. COMPUTED TOMOGRAPHY

The use of computed tomography (CT) in fetal (OE) is not recommended due to the effects of radiation, particularly in the first two trimester [21]. Postnatally, CT has its value because the presentation of bony defect is excellent [2]. However, CT is less effective than magnetic resonance imaging (MRI) in 'depicting soft tissue components in OE' [21]. A better assessment of the content can be achieved using water-soluble contrast material. CT cisternography scanning is beneficial in demonstrating communication between encephalocele and the intracranial subarachnoid space.

8. MAGNETIC RESONANCE IMAGING

Prenatal MRI exploration in the diagnosis of OE and other congenital anomalies is effective and despite the lack of maternal and fetal sedation, there are fewer limitations compared to the ultrasound techniques [21, 9] (Fig. 4). Postnatally, MRI is the method of choice in the diagnosis of OE. The method includes T1 and T2-weighted sequences possessing ability to depict brain tissue in multiple projections, to present cranial defects of the skull and to illustrate the

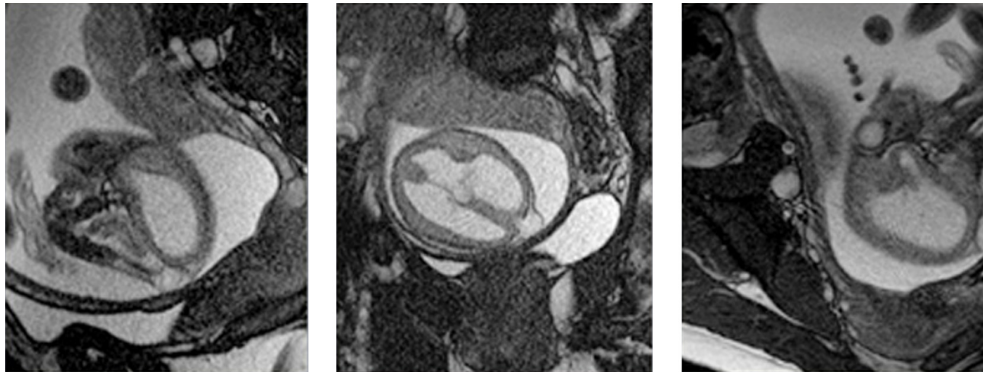


Fig. (4). Fetal occipital encephalocele, multiplanar T2W fetal MR imaging (25. gestational week) of occipital cephalocele associated with Chiari II malformation- hydrocephalus, small crowded posterior fossa and CSF-filled cephalocele extending through the dorsal skull defect. (A higher resolution / colour version of this figure is available in the electronic copy of the article).

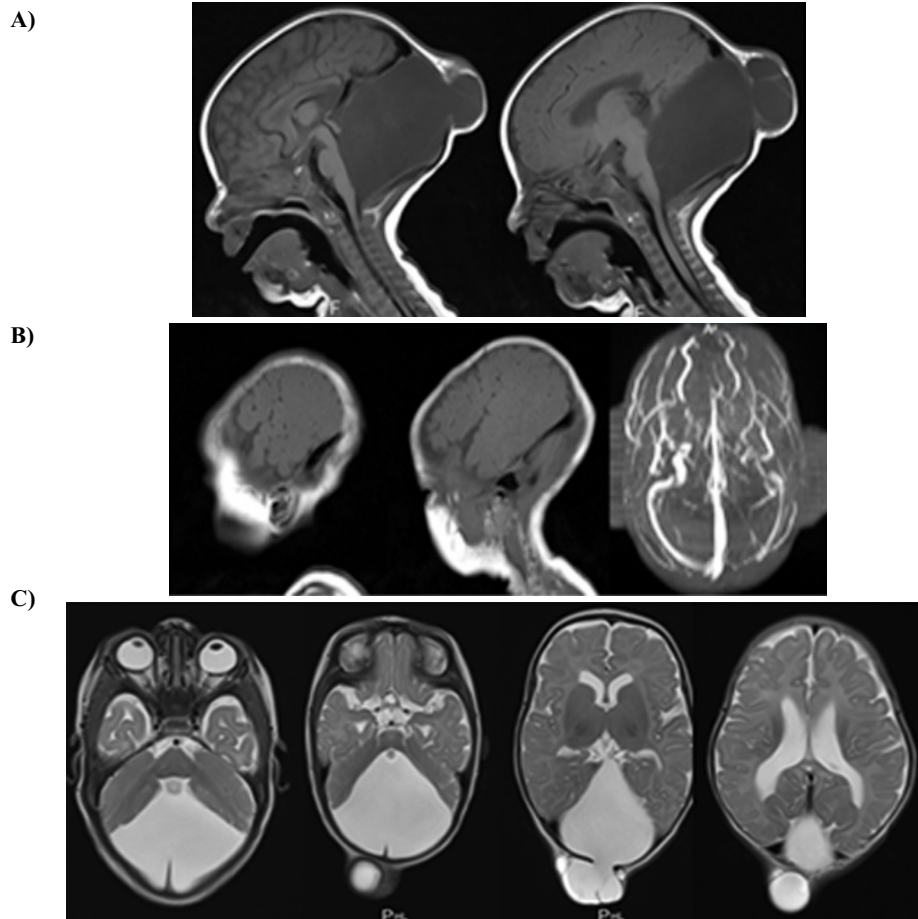


Fig (5). Occipital cephalocele associated with a Dandy-Walker cyst **A,B,C**): sagittal T1W and axial T2W images show big fourth ventricular cyst, expanded posterior fossa, significantly elevated tentorium and posterior bulge due to herniated CSF-filled sac extending through the occipital calvarial defect. (A higher resolution / colour version of this figure is available in the electronic copy of the article).

extent of the herniated cerebral tissue in the sac. The demonstration of other intracranial anomalies (Arnold-Chiari malformation, holoprosencephaly, Dandy-Walker complex, aqueduct stenosis, agenesis of the corpus callosum) is also possible and more useful in their analysis than CT (Figs. 1,2,3 and 5). MRI coupled with magnetic resonance angiography (MRA) is a beneficial method in the visualization of the contents of the sac and its relationship to venous sinuses [22]. MRI can usually depict the dural venous anatomy, and with MRI venography additionally improving the interpretation of the relationship between OE and venous sinuses, MRI

represents a superior method in the diagnosis of dilated venous sinuses [2, 19, 21] (Figs. 1,2,5).

9. TREATMENT AND PROGNOSIS

Surgical intervention is the best option for treating OE, and the most optimal time for surgery is between birth and 4 months. If the skin layer covers and protects the lesion, the intervention may be delayed within a few months; otherwise, without skin protection, the surgery should be done as quickly as possible [6]. The main goal of surgery is to

reposition the dislocated part of the brain tissue, meninges and CSF in the skull without causing neurological deficits, reparation of dural defect and cranioplasty [6]. The amount of brain tissue in the sac of OE, the state of CSF pathways, the degree of neurological deficit of the patients, and the existence (or non-existence) of the associated congenital malformation elsewhere in the body are the relevant factors for the management of the newborns with this lesion [2]. Surgical indication involves several conditions and should be considered in cases where there is a risk of rupture of the bag, leakage of CSF, meningitis, the content of prolapsed brain tissue, the presence of venous sinus, hydrocephalus, associated brain anomalies and aesthetic-cosmetic appearance [4, 19]. Dysplastic, gliosed, non-functional brain tissue should be excised, taking care of the essential vascular and brain structures [6, 11, 17, 19]. Lesion of torcula may provoke cerebral deep venous thrombosis. In neonates with microcephaly, there may be difficulty in the reposition of the prolapsed brain parenchyma and transverse sinuses, which might cause the occurrence of postoperative hydrocephalus [4, 19].

Cranioplasty is the final step in the surgical management of the OE, although not always necessary due to the osteogenic properties of the dura [11, 22]. Usually, a larger OE has a wider bone defect, while sometimes a smaller opening of the bone contains a greater content of intracranial elements [17]. A large bone defect requires urgent surgical intervention to avoid damage to the vital brain tissue as well as the surrounding blood vessels that penetrate the sack. A lesion of the blood vessels can result in a brain infarction [17].

Amongst newborns with OE, 60-70 % of the patients are at risk of developing hydrocephalus, which should be treated by application of the ventriculoperitoneal (VP) shunt [11, 17, 19]. In the majority of cases, hydrocephalus develops postoperatively. One of the mechanisms causing hydrocephalus may be torsion of the aqueduct of Sylvius or aqueductal stenosis. Surgical repair of the OE sac may provoke hydrocephalus due to changes in the CSF dynamics [19].

Inborn neurological deficits caused by this congenital disorder will not be improved by surgical intervention. Occasionally, multiple corrective interventions for the treatment of the accompanying anomalies of OE are also necessary [23].

The surgery of the giant OE with a large bony defect and prolapse of the significant brain elements into the sac is a highly- risky procedure. Extensive cranioplasty is a recommended method in protecting the functional brain parenchyma [5]. Microcephalic OE patients, with the sac containing cerebrum, cerebellum, and brain stem structures, have a poor prognosis regardless of the surgical intervention [5]. The giant OE sac may contain a large amount of brain parenchyma, sometimes occipital lobe and the brainstem, which can represent a significant challenge for surgical repair, especially if large venous structures (sinus torcular and transverse sinus) are closely connected with the sac [5]. Overall, patients with a giant OE have a poor prognosis [7].

10. PROGNOSIS

Many factors are relevant for evaluating the outcome in a patient with OE, such as the size of malformation, the amount and type of the dysfunctional brain elements in the sac (brainstem, the occipital lobes with or without the dural sinus) and the presence of hydrocephalus [5]. Patient's outcomes inversely correlate with the amount of brain parenchyma in the sac. Newborns with gigantic OE and a significant amount of brain tissue in a bag that could not be repaired without the risk of producing neurological deficit have a poor prognosis and there is poor confidence in the prediction of the length of the child's life [17]. In this case, the operation will, however, enable breastfeeding [6]. Hydrocephalus and microcephaly are poor prognostic factors associated with developmental delays and should be treated with expansible cranioplasty and VP shunt [17]. Seizures reported in at least 15% of the cases may additionally affect the quality of life [17].

11. MORBIDITY AND MORTALITY

Overall morbidity and mortality in patients with OE are still high in spite of advanced surgical management, and have varied significantly from 55% to 83% [1]. They have been improved in recent years, thanks to sophisticated high-resolution imaging, adequate and proper surgical treatment and adequate post-operative care. In a study of selective cases with OE in which patients with other malformations, with large lesions, with a significant amount of cerebral tissue in the sac that could not be repaired without accompanying risks, and with microcephaly were excluded, mortality was registered at only 2% [17]. Only six infants had some postoperative complications, four of which were amenable to treatment. In another study [24], no mortality of encephalocele was observed in nine years of follow-up.

A multidisciplinary approach and careful monitoring will improve the quality of life [1], and a number of specialists are often needed (neurologists, pediatricians, neurosurgeons, plastic surgeons and anesthesiologists) in proper preoperative, surgical and postoperative procedures [1]. A synergistic healthcare approach, including different types of medical, social, and professional services, is required to cater for these patients [3].

CONCLUSION

Occipital encephalocele is the most common form of the encephalocele and is manifested as swelling of different sizes over the occipital bone in the midline. The diagnosis is mostly based on the use of neuroimaging techniques. Operation is the best option for treatment and the proper time is between birth and 4 months. Overall, morbidity and mortality are still high in spite of advanced surgical management but have been significantly improved in recent years, thanks to sophisticated high-resolution imaging, adequate and proper surgical treatment and adequate post-operative care.

CONSENT FOR PUBLICATION

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CONFLICT OF INTEREST

The authors declare no conflict of interest, financial or otherwise.

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