Case Report

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Critical Care Obstetrics & Gynecology
ISSN 2471-9803

2021
Vol. 7 No. 6: 45

The Role Of Prenatal Ultrasound in Diagnosis of Fetal Encephalocele in Salmaneya Medical Complex: A 4 Years Retrospective Study With Literature Review

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Citation: Buhusayyen H, Hassani A, Mahmood N (2021) The Role Of Prenatal Ultrasound in Diagnosis of Fetal Encephalocele in Salmaneya Medical Complex: A 4 Years Retrospective Study With Literature Review. Crit Care Obst Gyne. Vol. 7 No. 6: 45.

Abstract

Encephalocele is characterized by a defect in cranial bone that results in protruding of the meninges (meningocele) or the meninges and the brain tissue (Encephalocele). Prenatal diagnosis can reveal a cranial defect with cerebral contents herniation of varying degrees, which are either purely cystic or may contain echoes of brain tissue. This paper focus on the encephalocele, prenatal diagnosis, associated syndromes as well as its management.

Keywords: Encephalocele; Prenatal diagnosis; Congenital anomaly

Received: August 11, 2021, **Accepted:** August 30, 2021, **Published:** September 30, 2021

Introduction

Encephalocele is a congenital malformation associated with a protrusion of the brain tissue and/or meninges through a mesodermal defect in the cranium and dura mater. It is one of serious neural congenital anomalies and its prevalence is estimated to be 0.8-5 per 10000 live births and it represents 15%-20% of all neural tube defects NTDs [1-4]. Encephaloceles commonly occur at the occipital and anterior region while midline encephalocele is rare. The incidences of certain types of encephalocele differ geographically and racially. Anterior encephalocele is more common in Asia, Africa and Russia while posterior type in Western Europe and South America. It may consist of meninges (meningocele), brain tissue with meninges (meningoencephalocele) or may communicate with the ventricles (meningoencephalocystocele) [5]. We report seven cases of frontal, parietal and occipital encephalocele.

The aim of study highlight the prenatal diagnosis of such neurological defects early and the role of multidisplinary team involvement in managing such cases that earlier termination of pregnancy is prohibited by county law. This study also provides an opportunity to identify the associated neural tube defects with encephalocele.

Material and Methods

From Jan 2018 till April 2021 the medical records with fetal encephalocele were reviewed. The cases were collected from the hospital records retrospectively. All newborns diagnosed with encephalocele have been reviewed for gestation history, folic acid consumption, gestational age, prenatal diagnosis, associated congenital anomalies and syndromes. Also, medical and surgical interventions were reviewed as well as postoperative follow up. All parents for affected patient were called for thorough evaluation for any neurodevelopmental delay. Verbal consents were obtained from all families [6-9].

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In a 4 years period, 7 patients were diagnosed with encephalocele (**Table 1**). Among 7 patients, four out of seven were diagnosed antenatal.

The diagnoses were made by 2D US. Three out of seven were above 35 years old, and 6 with paternal age above 35 years old. Medical history informed that 5 mothers used a folic acid (FA) tablets containing 5 mg, once daily after confirmation of pregnancy [10]. Neither started preconceptionally, nor consumed regularly. The rest didn't receive any supplements. Five mothers were Bahrainis while one was Pakistani and one was Philippine. Two of the couples were second degree relatives, Case-6 are cousins from both parents side, the others have no close consanguinity. One mother has previous three children with Meckel Gruber syndrome. None of the mothers used any teratogenic drugs or exposed to any radiation during pregnancy [11-15]. However one mother had received chemotherapy 8 months prior to gestation due to colon cancer. None were diabetic. None of the babies were product of Assisted Productive Technology (ART). Two were born

through vaginal delivery and five were born by cesarean section, one elective, four emergencies, one face presentation. The types of encephalocele were five occipital encephalocele, one parietal, and one frontal. The size varied from 2 cm × 2.4 cm-14.4 cm × 9 cm. Furthermore, four out of seven associated with dysmorphic feature, two associate with small Patent Foramen Oval (PFO), two with Patent Ductus Arteriosus (PDA), one of the PDA a small, Ventroseptal Defect (VSD) also detected by Echocardiogram. Case-4 was part of Walker Walburg syndrome and Case-6 associated with Mickel Gruber syndrome. Two cases of encephalocele died, Case-5 were encephalocele associated with anencephaly died few hours after the delivery, where Case-7 died a few days after the delivery due to lethal congenital malformation Meckel Gruber syndrome **(Figure 1)**. Case 2 and 4 were having convulsions and a significant neurodevelopmental delay during the follow up. There were no terminations of pregnancy for the prenatal diagnosed encephalocele due to religious cause.

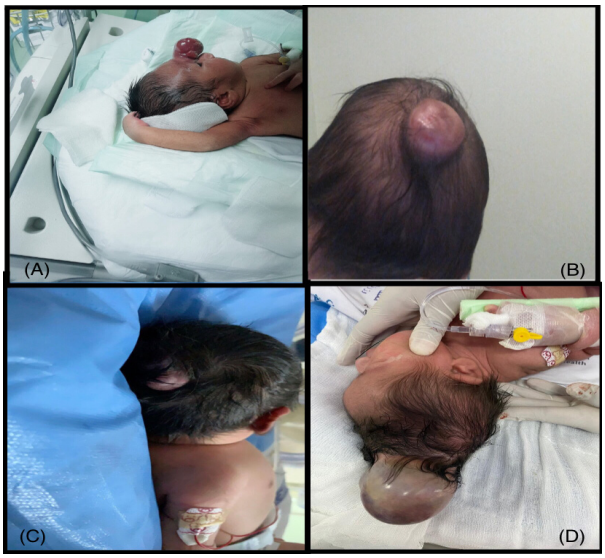

Figure 1 Neonates with different types of encephalocele; (A): Frontoethmoidal; (B,C): Occipital; (D): Parietal.

Discussion

Encephalocele may be congenital or acquired from trauma, tumour or iatrogenic injury. The risk factors vary widely which

Table 1: The demographic data and types of encephalocele.

	Case 1	Case 2	Case 3	Case 4	Case 5	Case 6	Case 7
Gravidity/Parity	G3P2	G6P4	G4P1A2	G5P4L3	G2P1	G4P3L1	G1P0
Gestational Age	39	36	35	37	34	35	37
Maternal Age	39	38	34	31	38	32	22
Paternal Age	42	41	45	35	36	43	29
Consanguinity	No	No	No	Yes	No	Yes	No
Maternal Comorbodities	Breast and colon CA, Hypothyrodism	B-thalassemia trait, G6PD reduced activity	Pregnancy induced hypertension	Nil	Nil	SCD	Nil
Nationality	Bahraini	Bahraini	Bahraini	Bahraini	Philippine	Bahraini	Pakistani
F.A Consumption	Yes	Yes	No	Yes	Yes	Yes	No
Prenatal Diagosis	Yes	Yes	No	No	Yes	Yes	No
Performer of Anatomy Scan	FM consultant	FM consultant	Registrar	Registrar	FM consultant	FM consultant	General practioner
Sex	Female	Female	Male	Male	Female	Male	Male
APGAR Score	9,10,10	1,5,10	Not mentioned	9,9,10	3,4,4	4,6,8	8,10,10
Birth Weight	4.180	3.37	3	2.7	2	2.290	2.775
Encephalocele Type	Parietal	Parieto-occipital	Occipital	Occipital	Occipital	Occipital	Frontal
Encephalocele Size	6.6 x 6.3 x 4.8 (cm)	14.4 x 2.7 x 9 (cm)	2 x 2.3 x 2.4 (cm)	3.3 x 2 x 2.5 (cm)	N/A	10 x 8 (cm)	3 x 5 (cm)
MRI/US Findings (neonatal)	Holoprosencephaly, partial agenesis of the corpus callosum, and Dandy- Walker malformation associated with posterior fossa crowding and tonsillar herniation	corpus callosum agenesis, generalized brain edema with interruption of the myelination. In addition, there is mild tonsillar ectopia	bilateral asymmetrical supratentorial ventriculomegaly	semilobar holoprosencephaly, microcephaly, midline and posterior fossa structural abnormalities with lissencephaly. Also there were corpus callosum agenesis and basal ganglia abnormalities	Anencephaly	-	Large defect in frontal bone with herniation of right frontal lobe with surrounding CSF leak fluid

Associated Anomalies	small PFO	Dysmorphic feature, microcephaly, small VSD, small PDA, normal abdominal ultrasound	Mild PPS	Dysmorphic feature, low set ears, rocker bottom deformity, hypospadias, hypoplastic optic disc, retinochoroidal coloboma stage 3 and hypopigmented fundus	Nil	Dysmorphic, hydrocephalus, small rib cage, abdominal distension and palpable kidneys as well as bilateral talipus	Dysmorphic, Small PDA, small PFO, normal ultrasound abdomen
Outcome	Surgical repair, currently not on medications, no convulsions	Surgical repair, neurodevelopmental delay	Surgical repair, Normal milestone	Sever neurodevelopmental delay	Died after 2 hours	Died after few days	Surgical repair, Normal milestone
Associated Syndrome	-	-	-	Walker Walburg syndrome	-	Meckel Gruber Syndrome	-

may include; genetic, environmental, parental consanguinity, previous child with NTDs and TORCH infections (toxoplasmosis, rubella, cytomegalovirus and herpes simplex virus). Also 50% may be associated with chromosomal abnormalities.

Many theories explain the development of encephalocele and the most accepted theory is incomplete separation of the ectoderm from neuroectoderm after closure of neural folds. Usually the cranial neuropore closes on day 25 of embryogenesis. Any abnormality that happens before this period may result in encephalocele [9,10].

Encephalocele classified according to the site of the defect into; anterior which is between the bregma and the ethmoid bone, occipital which is between the foramen magnum and lambdoid suture and finally parietal which is between the lambdoid suture and the bregma bone. Furthermore the anterior encephalocele is subdivided into frontal lesions that arise near the root of the nose and the basal lesions within the nose and the pharynx or the orbit. Occipital encephalocele is the most common form.

Furthermore, it associated with syndromes such as: Meckel-Gruber syndrome, Goldenhar's syndrome, Walker Warburg syndrome, Von Voss-Cherstvoy syndrome and it is also seen as a part of Amniotic band syndrome, Fraser syndrome, Roberts's syndrome [16-19].

In our study one case was associated with Dandy walker malformations, and one had walker Warburg syndrome and one with Mickel Gruber syndrome. Prenatal diagnosis of encephalocele can be achieved by measuring the level of maternal alpha fetoprotein in combination with antenatal ultrasound.

Prenatal diagnosis of encephalocele using 2D ultrasound (2D US) has been introduced since 1992, the diagnosis can be made by 2D US **(Figure 2)**. Nearly 80% of encephalocele can be detected by 2D US [13]. It appears as a defect in the skull which contains a cystic or solid mass with a gyral pattern that is adjacent to the brain. Cranial ossification starts approximately at 10 weeks of gestation, and hence the diagnosis of encephalocele cannot be achieved before that age. Differential diagnosis includes cystic hygroma, teratoma, branchial cleft cyst, scalp edema, hemangioma, Mackle Gruber syndrome, and Chiari III malformation [14]. However detection rate depends on the performer skills as in our institution, only 3 out of 7 patient encephalocele were detected

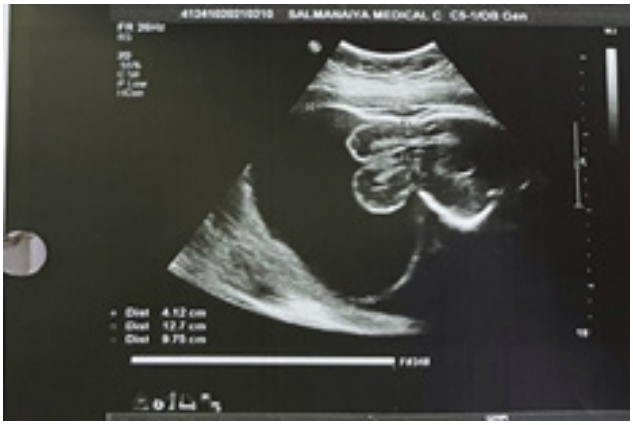

Figure 2 Transverse view of fetal head using 2D US showing defect in the calvarium with herniation of brain tissue.

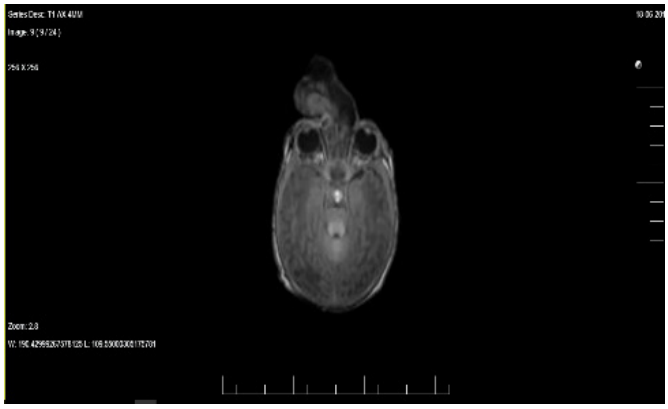

Figure 3 MRI brain with contrast reveal a right -sided defect in the nasal process of the frontal bone with herniation of brain parenchyma with CSF leak.

antenatal.

Moreover decrease folic acid intake Preconception and in early pregnancy has been linked to neural tube defect **(Figure 3)**. Amarin zo and colleagues recently reported that folic acid

fortified food reduce the incidence of neural tube defects including encephalocele. Although among our cases 4 out of 7 consumed folic acid postconception, none of the patients used preconception, 2 of them were on regular supplementation and 3 did not use any FA, which may be a predisposing factor for developing encephalocele.

Rowland and researchers reported that over 167 case, 70% were livebirths, 76% were isolated, detected in female infants and those with weight less that 2.5 kg. Newborn weighing more 3.5 kg had lower prevalence rate, while prevalence not affected by maternal age or plurality. In our study 4 were males and the rest were females, three of the women above 35 years old and 2 of them grand multipara and 6 of the fathers were above 35 years old. We cannot estimate the prevalence of encephalocele as there are other hospitals (private and governmental) in our country. The prognosis depends on; the size, site, the extent of the herniated tissue in the sac, hydrocephalus and the presence of associated anomalies. Lober and members found that approximately 50% of fetuses with encephalocele survive till birth. Most survivors with encephalocele have cognitive deficits, hydrocephalus, neurological deficits and seizure. In comparison to our study, two newborns died, two with normal milestone and 3 with sever psychomotor developmental delay and seizure.

Also, management of prenatal diagnosis of encephalocele is either by termination of pregnancy or continuation of pregnancy with multidipilinary team **(Figure 4)**. Which includes feta-maternal obstetricians, neonatologists, geneticists and neurosurgeons for surgical repair [16]. However, under Bahraini law, termination of pregnancy is forbidden. Hence, prenatal diagnosis has an eminent role in the management of such cases and it gives adequate time for psychological counseling of the parents, in addition to building management strategy. That is crucially depends on the site, size, contents, CSF leak, associated neurological involvements and other associated anomalies. Postoperative follow up should be on regular basis to detect of hydrocephalus and any neurodevelopmental delay.

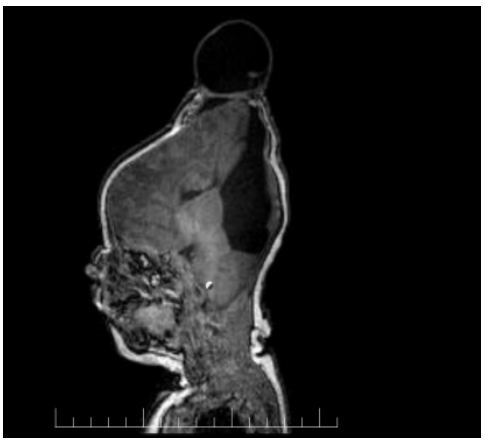

Figure 4 Sagittal view of MRI showing cranial meningoencephalocele in the high parietal region.

Conclusion

We present 7 cases of parietal, parietocciptal and occipital encephalocele which were detected prenatally using 2D US. Approximately 80% of cases can be detected in the first trimester with higher detection rate in the second trimester [13]. Detection of fetal anomalies helps in reducing the morbidity to the mother and the fetus, and allows planning the strategy.

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