



Prenatal Ultrasonographic and Magnetic Resonance Imaging Diagnosis of Occipital Meningo-Encephalocele- An Interesting Case Report

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ABSTRACT

Meningo encephalocele is a congenital anomaly and is a neural tube defect with occipital meningo encephalocele being the most common and is a result of a failure of the surface ectoderm to separate from the neuroectoderm. This condition can be identified in 1st trimester in 80% of cases and almost all by 2nd trimester. A 20-year-old third gravida was referred for antenatal Ultrasonography at five months of amenorrhoea to rule out fetal anomalies. On targeted imaging, for fetal anomalies, a defect was seen in occipital bone with herniation of posterior fossa contents with overlying meningeal covering. No other fetal anomalies were noted. A diagnosis of isolated occipital meningoencephalocele was made with additional fetal MRI correlation. The mother underwent termination of her pregnancy by Department of Obstetrics and Gynecology because of the grim fetal prognosis. The mother was advised to plan the subsequent pregnancies and was advised pre-conceptional folic acid supplementation. We present a case of isolated occipital meningoencephalocele- a rare congenital anomaly which was diagnosed prenatally in our hospital. This case provides an opportunity for identifying such neurological defects early and prompt termination of pregnancy to prevent comorbidity to mother. This study also helps to establish occipital meningoencephalocele as an isolated clinicoradiological diagnosis and to distinguish it from syndrome associated occipital meningoencephalocele or those associated with other neural tube defects like Chiari III malformations. It also allows us to stress once again the role of periconceptional folic acid in preventing the occurrence of neural tube defects.

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INTRODUCTION

Encephalocele is one of the most severe congenital neural tube defect. It occurs as a result of the failure of separation of surface ectoderm from neuroectoderm leading to a bony defect in the skull (Ugras and Kavak, 2016) characterized by a protrusion of brain and meninges through the defect. Prevalence of 0.8 – 5 per 10,000 live births is seen (Liao *et al.*, 2012).

Occipital encephalocele is the most common type of encephalocele with approximately 90% occurring in the midline and most of the cases are diagnosed prenatally. 60% of patients are seen associated with other malformations and chromosomal

defects (Ugras and Kavak, 2016). Serum alpha-fetoprotein and ultrasound establishes the prenatal diagnosis, and the prognosis depends on the extent of the herniated neural tissues and associated anomalies (Liao et al., 2012). Many times serum alpha-fetoprotein may remain normal because of intact meninges. Here is a report of one such illustrative case of occipital encephalocele.

CASE REPORT

A 20-year-old third gravida was referred for antenatal USG to rule out fetal anomalies at 21 weeks six days. The present conception was confirmed by first trimester dating scan at seven weeks, and the patient was asked to come for follow up scan at 11 weeks for assessing Nuchal translucency and gross fetal anomalies. Due to the prevailing COVID, 19 pandemic patient was unable to come for her 11-14 wks USG scan and instead came at 21 weeks for fetal anomaly scan. She could perceive fetal movements well. Past obstetric history - She has one living child; her second conception ended in a first-trimester early pregnancy loss. She had a significant medical history of hypothyroidism for six months and was on OralThyroid supplements (50microgram) with an euthyroid state at present.

Investigations

On Ultrasonography, all the fetal biometric parameters were corresponding to her clinical gestational age of 21 weeks five days. Targeted imaging for fetal anomalies revealed a defect of size 1.5 cm in the occipital bone (Figure 1 & Figure 3) with herniation of posterior fossa contents viz. both cerebellar hemispheres with overlying meningeal covering. The herniated cerebellum appeared partially atrophic with mild changes in contour and architecture with trans cerebellar diameter measuring 1.69 cm corresponding to 17 wks 6 days. Associated Bifrontal indentation with frontal bossing (Figure 2) of the fetal skull was also seen. Biparietal Diameter measured to 520 mm corresponding to the estimated fetal age of 21 weeks six days. No other fetal anomaly could be demonstrated. Craniovertebral junction and the fetal spine appeared normal. Fetal movements were noted with good cardiac activity (FHR - 158 bpm). With this a provisional diagnosis of occipital meningoencephalocele, fetal MRI was done to reassure the findings. On MRI, T2W sequence shows a T2 hyperintense cystic lesion in the posterior aspect of fetal skull communicating to the meningeal cavity through a defect in the fetal skull. The T2 hyperintense CSF containing meningeal cystic outpouching shows internal T2 signal intensities of that of brain parenchyma, resem-

bling partial cerebellar architecture along with the circular herophili (Figure 4). Brain stem and medulla appeared to be intact and within the cranial cavity. The craniovertebral junction could be separately identified with no cervical vertebral or spinal dysraphism (Figure 5). Since this is a congenital anomaly with a grim fetal prognosis, the patient was advised to undergo termination of pregnancy by the department of Obstetrics and Gynaecology.



Figure 1: Axial Sections were showing posterior calvarial bone defect with herniation of cerebellar contents.



Figure 2: Axial Sections were showing bifrontal indentation, causing frontal bossing of the fetal skull.

DISCUSSION

Encephalocele is a type of neural tube defect that occurs as a result of the failure of development between 25 and 27 days after conception (Franco et al., 2016).

It forms a part of a spectrum of neural tube defects that includes the following :



Figure 3: Sagittal view of fetal skull showing the posterior calvarial defect

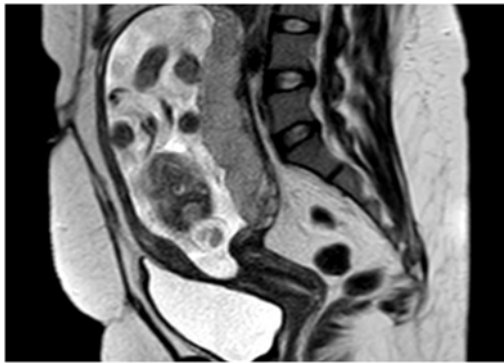


Figure 4: T2W MRI of the maternal pelvis in sagittal plane demonstrates an occipital calvarial defect showing herniated T2 hyperintense meningeal sac with T2 isointense posterior fossa contents likely cerebellum

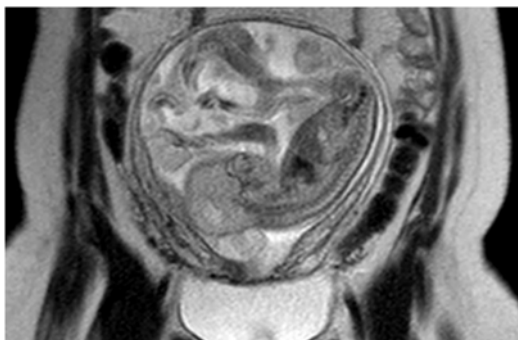


Figure 5: T2W MRI of the maternal pelvis in coronal plane demonstrates herniated sac with normal craniovertebral junction and fetal spine

1. Cephalocele is the term used for the protrusion of intracranial contents through a defect in the skull.
2. Meningoencephaloceles are the Cephaloceleles that contain herniations of brain tissue, meninges and CSF (Osborn *et al.*, 2012).
3. Meningocele refers to herniation of meninges and accompanying CSF without brain tissue. Atretic Cephaloceleles refers to a small defect that contains dura, fibrous tissue and degenerated brain tissue (Osborn *et al.*, 2012).
4. Gliocele refers to herniation of glia-lined pouch containing only CSF (Osborn *et al.*, 2012).

Encephaloceleles can be congenital or acquired and are classified by location and are named accordingly to the bone through which they herniate. They can be open or skin covered, and most of the congenital Encephaloceleles have coexisting intracranial abnormalities of varying severity (Osborn *et al.*, 2012).

Four most common forms of encephaloceleles are seen: sincipital (frontoethmoidal), parietal, occipital and skull base (Osborn *et al.*, 2012).

Encephaloceleles are multifactorial where environmental and genetic factors play a role. Folate deficiency is also associated with the pathogenesis of meningoencephaloceleles (Alwahab *et al.*, 2017).

The most common form of congenital encephalocelele is occipital encephalocelele (Franco *et al.*, 2016).

Occipital encephaloceleles can be diagnosed on ultrasound from about nine weeks. After cranial ossification at ten weeks, the skull defect and occipital sac can also be demonstrated, and in 30% of cases associated Spina bifida seen (Chougule *et al.*, 2006), and almost up to 80% of meningoencephaloceleles are detected during 1st trimester by Ultrasonography and by 2nd trimester almost all cases are identified (Liao *et al.*, 2012).

Demonstration of a skull defect with varying degrees of brain herniation is the basis for the diagnosis of encephalocelele on prenatal Ultrasonography (Ugras and Kavak, 2016).

On MRI T1 and T2 sequences are done, which shows characteristic signals of the brain and CSF (Franco *et al.*, 2016).

Occipital encephaloceleles are associated with syndromes such as

1. Meckel-Gruber syndrome which is characterized by broad nasal root, hypertelorism, cleft lip, cleft palate, microcephaly, microphthalmia,

polydactyly, polycystic kidneys and ambiguous genitalia (Liao *et al.*, 2012; Alwahab *et al.*, 2017; Ganapathy, 2014).

- Goldenhar's syndrome consists of a combination of facial macrosomia and auricular malformation associated with occipital encephaloceles (Ali *et al.*, 2019).
- Von Voss – Cherstvoy syndrome involves occipital encephalocele, thrombocytopenia, urogenital abnormalities and upper limb phocomelia (Franco *et al.*, 2016).
- Walker Warburg syndrome is congenital muscular dystrophy which is an autosomal recessive condition consisting of occipital encephalocele, myopia, retinal detachments, cataracts and aplasia (Franco *et al.*, 2016).
- It is also seen as a part of Fraser syndrome, Roberts's syndrome, and Amniotic band syndrome (Mahapatra, 2005).

Common Differential diagnosis Include Chiari III malformation, cystic hygroma, teratoma, branchial cleft cyst, scalp oedema, hemangioma and Meckel – Gruber syndrome.

In our case, a bony occipital defect was noted through which posterior fossa contents were seen herniating. The herniated contents included both the cerebellar hemispheres covered by intact meninges. The craniovertebral junction was normal. No obvious high cervical or other vertebral spinal anomalies were noted obviating the possibility of Chiari III malformation. No other fetal anomaly/ stigmata of syndrome association were demonstrable. Thus a diagnosis of isolated occipital meningoencephalocele was given.

CONCLUSION

This is a rare case of occipital meningoencephalocele associated with a low survival rate. Almost 80% of cases are identified in 1st trimester with almost all cases in 2nd trimester. 1st-trimester scanning of fetal anomalies helps to reduce the morbidity to the mother, and it also gives ample time for psychological counselling regarding expedited termination of pregnancy. In our case, the pregnancy was terminated, and the mother was advised to take pre-conceptional folic acid supplements before the next conception to avoid such congenital anomalies.

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Competing interests

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