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Case Report

From Cystic Sac to Smooth Cortex: Prenatal Imaging of an Occipital Encephalocele with Lissencephaly

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Abstract

Encephalocele is a rare congenital neural-tube defect characterized by herniation of meninges with or without brain tissue through a cranial defect. Lesions are classified by location-frontal, parietal, or occipital-the latter being the most frequent presentation. Prognosis depends on the anatomic site, the amount and integrity of herniated neural tissue, and the presence of associated abnormalities; when substantial brain is involved or major co-anomalies are present, outcomes are poor and pregnancy termination may be considered after counseling. We report a 23-week fetus with an occipital encephalocele identified on obstetric ultrasound; fetal MRI confirmed the diagnosis and, on targeted assessment for associated anomalies, demonstrated lissencephaly.

Keywords: *Occipital Encephalocele; Lissencephaly; US; MRI*

Introduction

Encephalocele is a cranial neural-tube defect in which meninges and, in true encephalocele, brain tissue herniates through a calvarial defect [1]. The condition is uncommon; most lesions in cohorts are posterior/occipital [1,2].

Imaging underpins antenatal detection and characterization: 2D ultrasound typically makes the diagnosis, with 3D ultrasound and fetal MRI detailing sac contents, venous anatomy, and associated brain anomalies to inform counseling and delivery planning [1-3]. Prognosis is largely determined by the volume/quality of herniated neural tissue and associated anomalies; management is multidisciplinary, with postnatal neurosurgical repair standard and in utero repair reported only in highly selected cases [1,4]. Here we report a 23-week occipital encephalocele associated with lissencephaly.

Case Report

A 30-year-old primigravida (G1P0) at 23 weeks' gestation was referred by her general practitioner after the routine second-trimester scan identified a posterior cystic mass suspicious for a cervical hygroma. She had no past medical or surgical history, was on no medications or supplements, reported no consanguinity, and had no family history of congenital anomalies. Routine maternal laboratory investigations were unremarkable.

A detailed ultrasound (Figure 1) demonstrated a large midline occipital, predominantly cystic, extracranial mass. A solid component within the sac showed echogenicity resembling brain parenchyma and maintained continuity with the intracranial compartment through a calvarial defect in the occipital bone, consistent with an occipital encephalocele. Color Doppler did not suggest a hypervascular solid tumor. Fetal biometry was appropriate for gestational age.

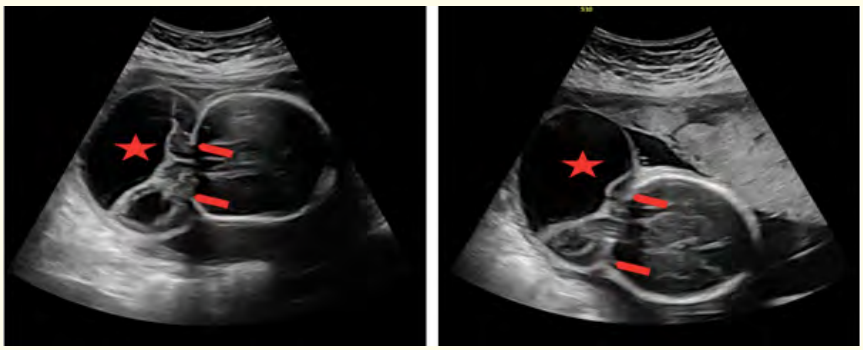

Figure 1: *Two axial fetal ultrasound sections showing a midline occipital, mainly cystic extracranial mass (asterisk) with an echogenic solid component continuous with intracranial brain through an occipital calvarial defect (arrows), diagnostic of an occipital encephalocele.*

Fetal MRI was performed for further characterization. Imaging (Figure 2) confirmed the occipital encephalocele with herniated neural tissue. In addition, the cerebral surface appeared abnormally smooth for gestational age, with absence of expected primary sulcation, findings compatible with lissencephaly. No extracranial non-neural malformations were identified on the sequences obtained.

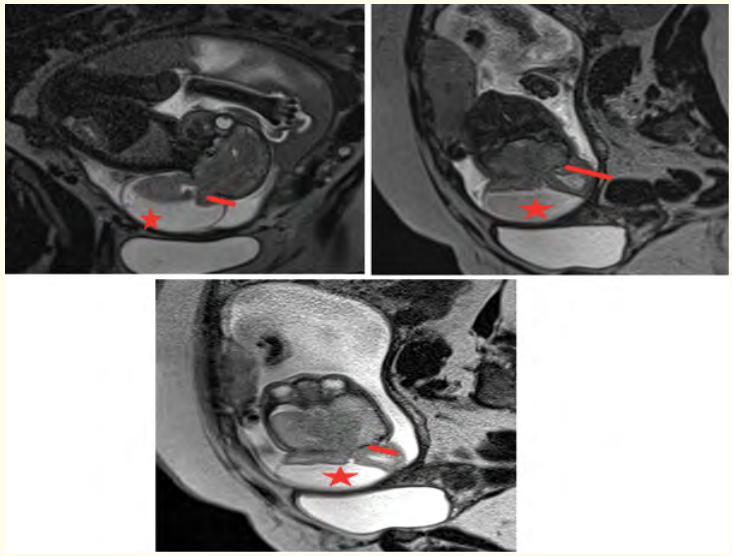

Figure 2: *Coronal, axial, and sagittal fetal MRI images show a midline occipital calvarial defect (arrows) with herniation of brain parenchyma and CSF forming a sac-consistent with an occipital encephalocele. In addition, the cerebral surface appears abnormally smooth for gestational age, with absent expected primary sulcation, compatible with lissencephaly.*

The patient was referred to clinical genetics and maternal-fetal medicine/gynecology for close follow-up and comprehensive counseling regarding prognosis, options for pregnancy management, and delivery planning. A plan for serial ultrasound surveillance and continued multidisciplinary care was established.

Discussion

A cephalocele is a herniation of meninges and/or brain through a cranial bony defect; the term encephalocele is used when the sac contains brain tissue, whereas a meningocele contains only cerebrospinal fluid. These lesions are typically skin-covered and are considered closed neural tube defects [1]. The estimated prevalence is on the order of 0.8-5 per 10,000 births. Geographic patterns exist: in Europe and North America, most lesions are occipital/posterior, while frontal (sincipital) lesions predominate in parts of Southern/Southeast Asia [1]. Reports from African settings describe relatively higher frequencies in some regions, linked to low socioeconomic status, consanguinity, limited antenatal care, and young maternal age; country-level observations include series from Central Africa and Morocco [5].

Encephalocele arises from a failure of cranial neural tube closure during the 3rd-4th weeks of embryogenesis, leaving a persistent calvarial defect through which meninges and/or brain may herniate [5]. Etiologies are heterogeneous, involving environmental/teratogenic exposures and genetic factors; for example, CEP290 variants have been associated with occipital encephalocele in some reports [5].

The classic sonographic appearance is a midline, sac-like mass in continuity with the cranial vault that communicates with the intracranial compartment through a calvarial defect; the sac is commonly skin-covered. Occipital lobes are the most frequent contents, and cerebellar tissue may be present in very low posterior lesions. Two-dimensional ultrasound detects a large majority of encephaloceles, typically by the second trimester (with first-trimester cases described), while 3D ultrasound improves depiction of the defect's location/extent and intracranial anatomy and aids counseling [1,2,5].

Fetal MRI provides high-contrast assessment of brain parenchyma and the posterior fossa when ultrasound identifies a cephalocele, and is valuable for detecting subtle intracranial abnormalities and guiding perinatal planning [1,7]. Advanced techniques such as diffusion tensor imaging can delineate white-matter tracts and demonstrate continuity between extracranial and intracranial tissue; susceptibility-based sequences help define venous anatomy [3].

Associated malformations are frequent: cerebellar dysplasia, corpus callosum agenesis/dysgenesis, Dandy-Walker malformation, Chiari spectrum, and craniofacial/extracranial anomalies [1,5]. MRI series also document malformations of cortical development such as polymicrogyria and neuronal heterotopia [3]. Syndromic associations include Walker-Warburg and Joubert spectrum disorders; because Walker-Warburg lies within the cobblestone cortical malformation spectrum, co-occurrence with lissencephaly-spectrum pathology is recognized in syndromic cases [1,3].

Midline scalp or craniofacial masses that can mimic encephalocele include dermoid/epidermoid cysts, hemangioma, cephalohematoma, cystic teratoma, cystic hygroma, branchial anomalies, scalp edema, and even prominent fetal hair late in gestation. The key discriminator is a calvarial defect with intracranial continuity; a systematic search for additional anomalies is essential once a defect is confirmed [1,7].

Evaluation should include a targeted search for associated anomalies and genetic testing when indicated; delivery planning is multidisciplinary. Postnatal neurosurgical repair aims at watertight dural closure with skull reconstruction, with individualized mode of delivery [1,4,5]. Prognosis is chiefly determined by the amount/quality of herniated neural tissue and by associated anomalies. Large posterior sacs with cerebrum/cerebellum or ventricles and hydrocephalus have worse outcomes and may require CSF shunting. Fetal repair has been reported only in small cohorts and remains investigational [1,4,5].

Conclusion

In summary, this 23-week case of occipital encephalocele with associated lissencephaly underscores the pivotal role of systematic prenatal imaging and multidisciplinary care. Ultrasound-augmented by three-dimensional acquisitions-flags the presence and configuration of the sac, whereas fetal MRI refines the assessment of extracranial neural tissue, cortical maturation, and concomitant brain anomalies that drive counseling and delivery planning.

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Conflict of Interest

The authors declare no conflict of interest.

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