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CASE REPORT PEER REVIEWED | OPEN ACCESS

Antenatal diagnosis and management of an encephalocele in Jamaica

Tracey Martinborough, Tiffany Hunter-Greaves, Peter Charles, Peter Johnson, Nadine Johnson

ABSTRACT

Introduction: Encephalocele is a rare fetal anomaly associated with neurodevelopmental morbidity, especially in developing countries where management resources are limited.

Case Report: Here, we present a case of antenatal diagnosis of an occipital encephalocele in a 25-year-old multigravida. Ultrasound and magnetic resonance imaging were used throughout the second and third trimesters to guide delivery and plan postnatal repair. A multidisciplinary approach to care, involving maternal–fetal medicine, radiology, neonatology, and neurosurgery specialties, was undertaken from mid-gestation. This multiteam approach afforded predelivery planning that resulted in an uneventful cesarean delivery and surgical correction of the encephalocele within 24 hours of extrauterine life.

Conclusion: This case demonstrates that even in the absence of advanced resources for fetal interventions, successful outcomes can be achieved with early diagnosis, a multiteam approach to care and predelivery planning.

Keywords: Encephalocele, Jamaica, Neural tube

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INTRODUCTION

Primary occipital encephalocele results from congenital defects in the posterior cranium leading to meningeal eversion through the defect [1]. Atypical expression of Hox genes in the cranial neural crest can lead to abnormal skeletogenesis and endochondral bone malformation in the occiput [2]. Despite having a strong association with chromosomal and genetic disorders, environmental risk factors are equally important determinants of isolated neural tube defects (NTDs) such as cephaloceles [3]. Open NTDs have been linked to folate deficiency, pregestational diabetes, and maternal obesity [4–6]. The increasing rates of obesity and pregestational diabetes combined with a lack of pre-pregnancy planning is likely to lead to an increase in these birth defects in Jamaica, mirroring the rise in developed countries [7, 8].

The current estimated prevalence of open NTDs in the Caribbean is 3.76 per 10,000 live births. This rate is significantly lower than that in other western countries, but it represents an important health concern due to the paucity of neonatal and pediatric neurosurgery resources

[9]. Open NTDs, such as encephaloceles, are associated with significant perinatal morbidity and mortality [10]. As evidenced in our case of antenatal diagnosis of an occipital encephalocele in a 25-year-old multigravida, we believe that increased uptake of universal mid-trimester screening and a multidisciplinary approach to antenatal care can improve outcomes in low-resource settings.

CASE REPORT

Maternal history

A 25-year-old gravida 2, para 1001 woman was referred to our unit for evaluation, counselling, and continued care after a routine anatomy survey performed at another institution indicated a large occipital encephalocele at 20 weeks and 5 days of gestation. The mother had no known chronic medical illnesses, a normal body mass index, and no family history of genetic syndromes. She had delivered a healthy male infant at term three years prior after an uneventful antenatal period. The index pregnancy was complicated by hyperemesis gravidarum, requiring multiple admissions in the early second trimester to a rural hospital not equipped with ultrasound services or aneuploidy screening. The mother had no history of maternal febrile illness or pesticide exposure. She was initiated on folic acid at pregnancy confirmation at 12 weeks of gestation at the time of pregnancy diagnosis. Laboratory investigations in the first and second trimester revealed normal findings. The patient had an ultrasound for anatomy survey at a private radiological service at 20 weeks and 5 days prompting referral to our tertiary level hospital.

A level 3 obstetric ultrasound performed at our unit at 27 weeks and 3 days of gestation revealed a live singleton with microcephaly, a defect measuring 1.2 cm in the occipital cranium, and an occipital encephalocele measuring 7.0 × 6.3 × 5.0 cm (Figure 1). The cerebellum and cisterna magna appeared hypoplastic with Chiari malformation (Figure 2). The cerebral ventricles, cavum septum pellucidum, and all other organ systems appeared normal.

The patient and her partner were counselled at the initial and subsequent visits on the associated risks of perinatal morbidity, poor prognosis, and perinatal mortality. Amniocentesis for G-banded karyotype and microarray were offered, but they were declined. Pregnancy termination was also offered; however, decision was made by the patient to continue the pregnancy. Therefore, multiteam management was commenced in collaboration with the neurosurgery and neonatology units. Subsequent fetal magnetic resonance imaging (MRI) and level 3 ultrasounds revealed interval increase in the size of the encephalocele, a small appearing posterior fossa, but no tonsillar herniation or other cranial anomalies (Figures 3, 4A and B). Normal abdominal circumference, femur length, umbilical artery Doppler study findings, and biophysical profile scores were obtained on serial growth scans throughout the third trimester. Preparations were made for an elective cesarean delivery at term; however, the patient presented in the active first stage of labor with membrane rupture at 36 weeks of gestation. An emergency classical cesarean section was performed with no complications. The post-cesarean course was uneventful, and the mother was discharged on the third postoperative day.

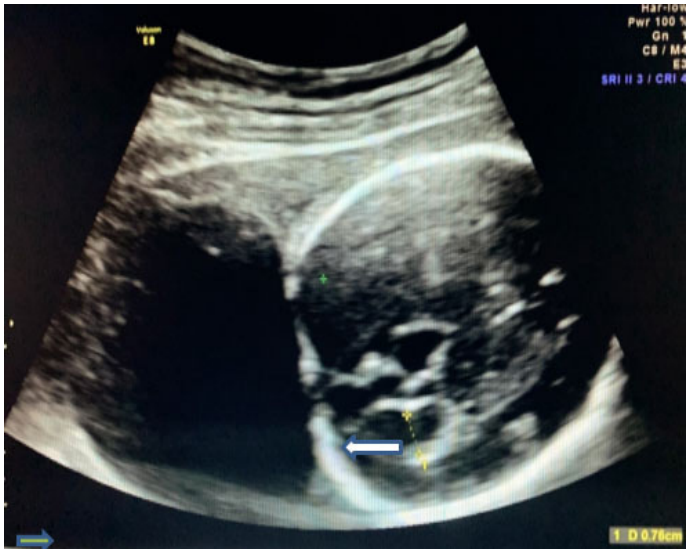

Figure 1: Cross-sectional view of the foetal cranium. The gray arrow indicates a thin-walled cystic lesion related to the occiput and upper posterior neck of the fetus measuring 7.0 cm (L) × 6.3 cm (W) × 5.0 cm (AP). The white arrow indicates a defect in the posterior cranium. A normal posterior–lateral ventricle is also seen.

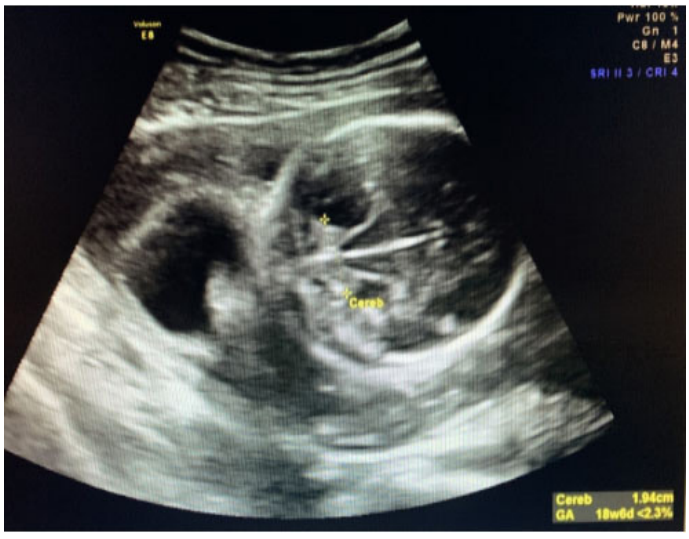

Figure 2: Cross-sectional view of the foetal cranium. The image demonstrates a hypoplastic cerebellum and cisterna magna.

Neonatal evaluation and follow-up

A female infant weighing 3230 grams, with APGAR scores of 9 at 1 minutes and 9 at 5 minutes, was delivered

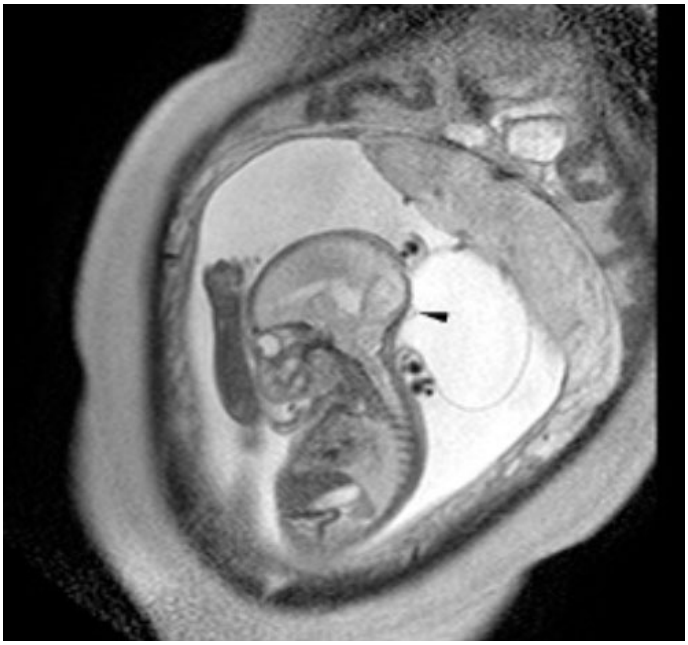

Figure 3: T2-weighted magnetic resonance image in the fetal sagittal plane demonstrates a thin-walled cystic lesion related to the occiput and upper posterior neck of the fetus measuring 8.5 cm (L) × 6.7 cm (W) × 6.5 cm (AP). A tiny defect in the occiput (arrowhead) with a tiny punctum can be observed.

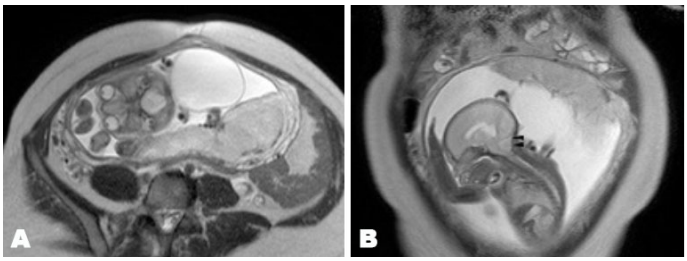

Figure 4: (A) Axial and (B) sagittal T2-weighted imaging demonstrating a small appearing posterior fossa, but no tonsillar herniation or other anomalies.

by cesarean section (Figures 5A and B). On day 1 of life, the female neonate underwent surgical correction of the encephalocele. There were no cerebral or cerebellar herniations, and primary closure of the cranial defect was performed using allopathic skin. A ventriculoperitoneal shunt was not placed, and the immediate postoperative period was uneventful. The infant was discharged to home on day 7 of life with continued neurosurgical follow-up.

Postnatal MRI was performed at three months of age and after closure of the encephalocele. The previously demonstrated occipital encephalocele was absent, consistent with the surgical history. There was a small midline defect in the occipital bone, representing the previous communication with the cephalocele (Figure 6A). The fourth ventricle was small, and a cerebrospinal fluid-filled tract extended from its right posterior margin through the superior aspect of the cerebellar vermis to the cerebellar surface (Figure 6B). The posterior fossa appeared small, with crowding of the cerebellum. No cerebellar tonsillar descent was noted. There was gross lateral and third ventriculomegaly, that appeared secondary to aqueductal stenosis. Aqueduct stenosis occurred secondary to distortion of the anatomy around the tentorium owing to crowding within the posterior fossa. Tectal beaking was noted. There was a prominent massa intermedia of the thalamus. The overall findings suggested a variant of Chiari II malformation. The infant continues to be followed closely by neurosurgery, pediatric neurology, and physiotherapy services as neurodevelopmental challenges are anticipated.

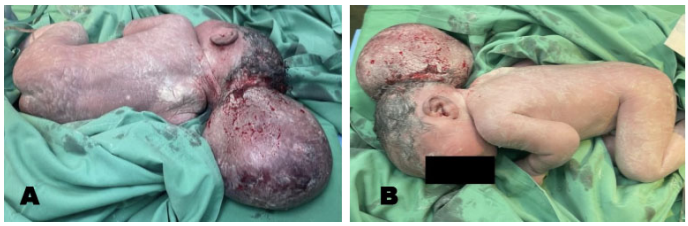

Figure 5: Gross images taken immediately after cesarean delivery demonstrating occipitoencephalocele measuring 9.5 cm (L) × 6.5 cm (W) × 6.5 cm, (A) posterior view and (B) anterior view.

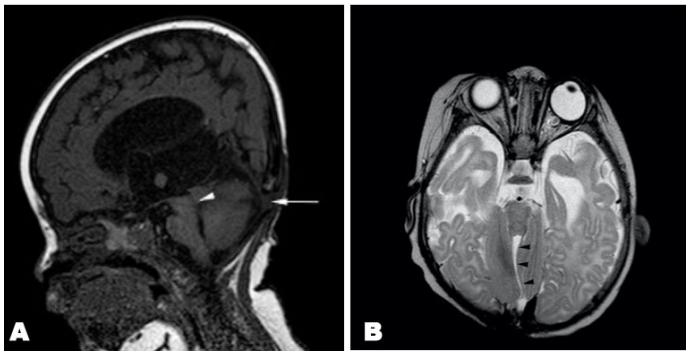

Figure 6: Sagittal and coronal T1-weighted imaging. Gross lateral and third ventriculomegaly with stenosis of the aqueduct of Sylvius (arrowhead). A small midline defect can be seen in the occipital bone. (A) A prominent massa intermedia is noticeable. (B) The fourth ventricle is small, with a cerebrospinal fluid-filled tract extending from it to the surface of the cerebellum (arrowheads).

DISCUSSION

Over the past decade, intrauterine fetal surgery has been the optimal management for antenatally detected open NTDs amenable to surgical correction [11]. Typically performed in specialized referral centers with first-world infrastructure and support services, these procedures have led to improved neurodevelopmental outcomes in the affected population. However, in settings unable to provide fetal surgical services, increased emphasis should

be placed on prevention with folic acid supplementation, timely antenatal diagnosis, and early postnatal surgical correction [12, 13]. Studies in populations with folic acid food fortification have shown that despite fortification, a considerable number of infants born to reproductive-aged women are still at risk of congenital anomalies, which underscores the need for additional antenatal supplementation [14]. Our patient resides in a country with folic acid fortification, but commenced oral supplementation later in the first trimester, likely after closure of the anterior and posterior cranial neuropores. Pre-pregnancy folic acid supplementation is recommended as neuropore closure occurs between 22 and 28 days after conception, typically before the missed menstrual period [15]. In addition to the need for supplementation and fortification, our case demonstrates the value of pregnancy screening, diagnosis of fetal NTDs, and multiteam management.

Early detection provides both healthcare providers and patients with the opportunity to undergo aneuploidy testing, pregnancy termination, or delivery planning to optimize outcomes. In Jamaica, aneuploidy testing is not widely available, and its cost can be prohibitive. Additionally, there is a general cultural antipathy toward termination of pregnancy, especially beyond the first trimester; underscoring the importance of early sonographic detection of fetal anomalies. First trimester ultrasound screening remains the cardinal recommendation in settings with follow-up diagnostic procedures and resources to perform antenatal surgical repair [16]. However, in low-resource settings like Jamaica, with a paucity of skilled sonographers, mid-gestation sonographic screening of fetal anatomical defects remains the standard practice.

In addition to a second-trimester screen, our patient benefitted from maternal–fetal medicine and radiology consultations with MRI verification of the anatomical defects. Antenatal imaging of fetal central nervous system abnormalities using MRI is a useful adjunct for the determination of candidacy for prognostication and surgical planning, whether fetal or post-delivery [17]. In our case, intrauterine MRI imaging guided repair planning even after delivery, as the same was not available in the immediate postpartum period. Combined with specialist imaging, antenatal neurosurgical consultation and planning permitted surgical correction of the defect within 24 h of extrauterine life. Multidisciplinary pregnancy management continued into the immediate postpartum period as this approach has been proven to improve neonatal outcomes in open NTDs [18].

However, the long-term prognosis in our case is uncertain; motor deficits, seizure disorder, and the development of hydrocephalus will be monitored via neurosurgical follow-up. Nonetheless, the progress made to date is not commonplace in our resource-restricted setting and reflects the advances made in antenatal detection, planning, and multidisciplinary management of encephaloceles and NTDs in general.

CONCLUSION

This case demonstrates that even in the absence of resources for fetal interventions, successful outcomes can be achieved with early diagnosis, a multiteam approach to care, and predelivery planning.

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Author Contributions

Tracey Martinborough – Conception of the work, Design of the work, Acquisition of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Tiffany Hunter-Greaves – Conception of the work, Design of the work, Acquisition of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Peter Charles – Conception of the work, Design of the work, Acquisition of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Peter Johnson – Conception of the work, Design of the work, Acquisition of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Nadine Johnson – Conception of the work, Design of the work, Acquisition of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission

The corresponding author is the guarantor of submission.

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Consent Statement

Written informed consent was obtained from the patient for publication of this article.

Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.