Fetal MRI for characterising a variety of posterior fossa anomalies suspected on 3rd trimester ultrasound examination – a short series of 4 cases

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Abstract
Fetal MRI is increasingly being used to more accurately assess abnormalities detected on screening ultrasound. The procedure is more pertinent when the initial ultrasound is done late in the third trimester and when the abnormality involves the posterior fossa of the brain. Four cases with a variety of unusual posterior fossa anomalies are presented.

Introduction
Prenatal ultrasound (US) is the investigation of choice when screening for fetal abnormalities. When an intracranial abnormality is detected on US specifically within the posterior fossa, a fetal MRI is the next investigation of choice, to accurately characterise and diagnose the abnormality. In the developing world, patients often present as unbooked pregnancies in the third trimester. At this late stage of pregnancy, the sensitivity of US decreases owing to ossification of the cranium and the relative size of the fetus compared with the volume of liquor, making fetal MRI more valuable.

We present four instances where posterior fossa abnormalities were suspected on third trimester antenatal US. Fetal MRI scans diagnosed four specific posterior fossa abnormalities including a Dandy-Walker malformation, a Joubert syndrome and related disorder, an occipital encephalocoele, and an occipital bone lesion.

Case reports
Antenatal US was performed for the first time during the third trimester in all the cases, owing to unbooked pregnancy, which is common in developing countries with poorly distributed resources. MRI scans on all patients were performed at a referral unit on a 1.5 Tesla machine (General Electric Signa Excite, Michigan, USA). Routine sequences included T2, T2* and T1.

Case 1
A 28-year-old woman, 32 weeks pregnant, presented to the antenatal clinic and was referred for antenatal US for suspected polyhydramnios. Antenatal US demonstrated polyhydramnios, an enlarged cisterna magna (>10 mm), a cavum septum pellucidum, and retarded long bone growth (at 5th percentile). There was no echogenic vermis visible but the lateral ventricles (7 mm) and aqueduct were normal. There were no features of spina bifida.

Fetal MRI (Fig. 1) confirmed the Dandy-Walker malformation on the basis of an absent vermis, cystic dilatation of the fourth ventricle, an enlarged posterior fossa, upward displacement of the torcula and splayed hypoplastic cerebellar hemispheres.[1] There was no hydrocephalus, corpus callosum agenesis, a normal pontine bulge, no intracranial cysts or spinal abnormalities. The patient was counselled and the child was delivered by caesarian section and found to be a trisomy 18. The child did not survive.

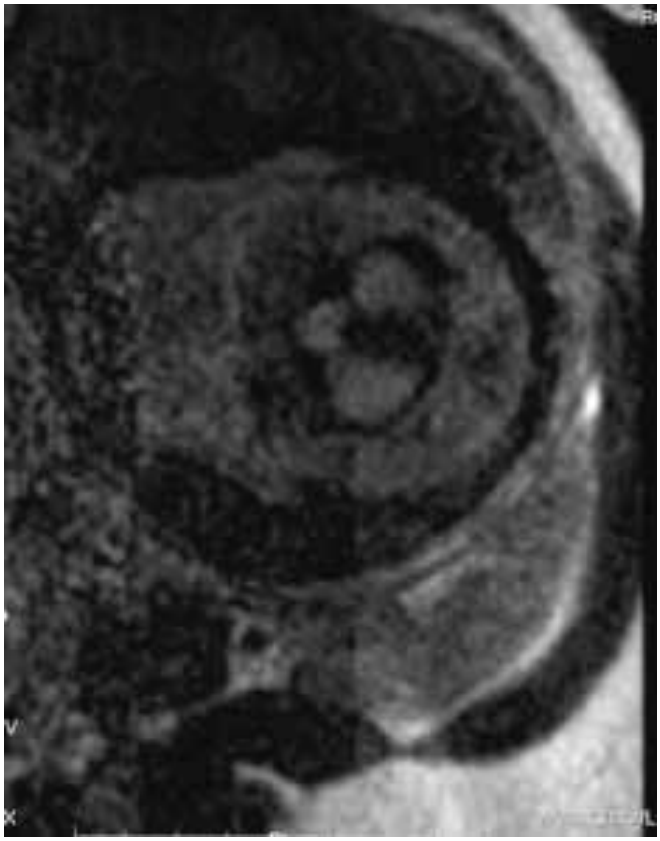

Fig. 1. Case 1: Axial fetal MRI: Splayed hypoplastic cerebellar lobes, and an enlarged cisterna magna and vermian agenesis consistent with a Dandy-Walker complex.

Case 2

A 30-year-old woman, 39 weeks pregnant, presented for a late third trimester US with a history of decreased fetal movements. Antenatal US demonstrated a possible occipital encephalocoele. The scan was considered suboptimal in view of the advanced gestational age.

Fetal MRI (Fig. 2) confirmed the encephalocoele by demonstrating continuity with the intracranial compartment and demonstrating brain tissue extracranially. The occipital bony defect of 3.6 cm was also demonstrated. There was no associated hydrocephalus, brain atrophy, spinal abnormality or other anomaly.

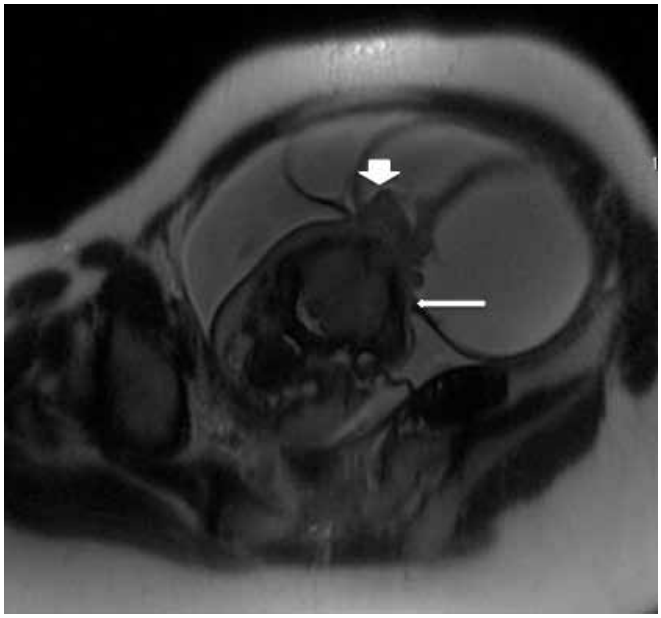

Fig. 2. Case 2: Axial fetal MRI: An occipital defect(long arrow) with large encephalocoele containing cerebellar tissue (arrow head) is demonstrated.

The child was delivered by caesarean section a week later and proved to be blind and spastic, and the prognosis was poor. Neurosurgical intervention was considered unfeasible.

Case 3

A 31-year-old woman who was 31 weeks pregnant presented at the antenatal clinic for US; this demonstrated hydrocephalus and an abnormal posterior fossa but did not provide a more specific diagnosis. On fetal MRI (Fig. 3), a diagnosis of Joubert syndrome and related disorders[2] was made by the pathognomic finding of a 'molar tooth' sign at the ponto-mesencephalic junction, representing vermian aplasia and stretched superior cerebellar peduncles with a deep interpeduncular fossa.

Case 4

A 37-year-old woman presented to the antenatal clinic at 28 weeks' gestation. Owing to her advanced maternal age, she was referred for an antenatal US. This demonstrated a cystic structure in the posterior fossa and possible dysplasia of the vermis. A Dandy-Walker complex was considered.

Fetal MRI (Fig. 4) showed a well-defined extra-axial lesion within the occipital bone. The vermis, cerebellar hemispheres and fourth ventricle were normal, therefore excluding a Dandy-Walker complex. The lesion was reported as either a haemangioma or a cyst of the occipital bone.

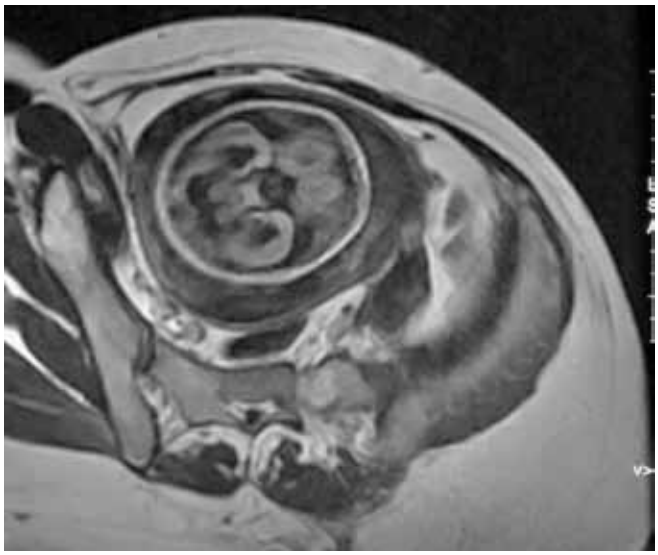

Fig. 3. Case 3: Axial fetal MRI: 'Molar tooth' sign consistent with Joubert syndrome and related disorders.

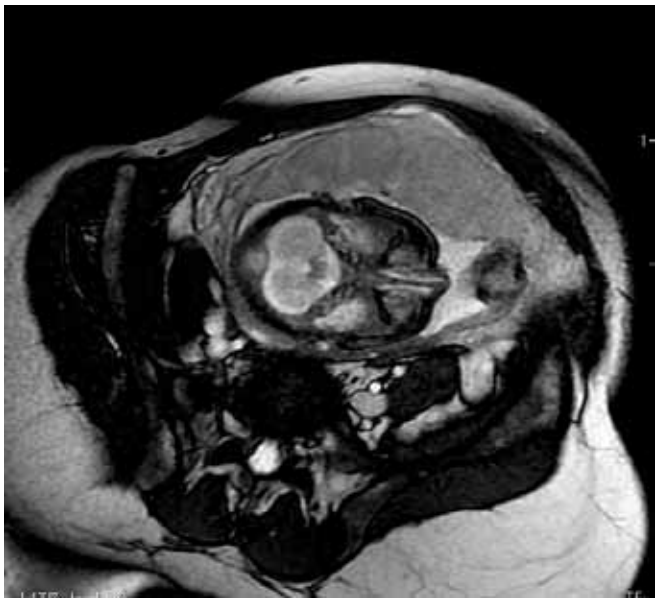

Fig. 4. Case 4: Axial fetal MRI: Well-defined extra-axial lesion situated within the occipital bone. The intracranial posterior fossa structures are normal.

Discussion

MRI is increasingly used to evaluate the fetal brain, particularly when an abnormality has been detected on prenatal US or when a fetus is at increased risk for neurodevelopmental anomalies. Fetal MRI has several advantages over US. It has higher contrast resolution, is not affected by the shadowing from the calvarium or by low amniotic fluid volume, allows a larger field of view and can easily be performed using ultrafast T2-W sequences,[3] making it more useful than US in late pregnancy. Parents also better understand the images and therefore counselling is made easier. Counselling of parents is important for obtaining appropriate knowledge of the potential outcomes in the neonate and child.[4] Disadvantages include limitations owing to fetal motion, poorer spatial resolution than US, cost, availability and lack of expertise. One of the primary reasons for performing fetal imaging is to gather accurate information about fetal structure on which to base reliable counseling. This parental counseling is important for making

informed decisions regarding future management of the pregnancy, regarding offering termination, planning delivery, post-natal prognosis and genetic counseling and testing.[4]

In terms of the posterior fossa specifically, fetal US has limited specificity, and false-positive diagnoses are well described. A recent study showed a 33% false-positive rate for prenatal US compared with MRI.[4] The same study, however, also highlighted the limitations in both sensitivity and specificity of fetal MRI compared with postnatal MRI. In cases of posterior fossa anomalies, only 60% of prenatal MRI diagnoses are confirmed postnatally. One of the main factors contributing to this low sensitivity and specificity is the poor spatial resolution for small structures such as the brainstem and vermis, especially in early gestation.[4] This emphasises the need to perform fetal MRI studies after 22weeks' gestation and the need for postnatal MRI to correlate fetal MRI findings.

The two cases of Dandy-Walker complex and Joubert syndrome and related disorders highlight the superiority of MRI in evaluating for vermian dysplasia or aplasia, the shape of the fourth ventricle, the insertion of the tentorium cerebelli and the presence of the pontine bulge. It is also essential to look for associated cerebral and extra-cerebral abnormalities when a posterior fossa anomaly has been detected; this additional information is more accurately obtained with fetal MRI. Associated migrational abnormalities and midline anomalies specifically, are more thoroughly assessed on MRI.[5] A posterior fossa cyst can severely hamper evaluation of the posterior fossa structures on ultrasound, as demonstrated in the fetus with the occipital encephalocele.

Conclusion

Our cases have highlighted the superiority of MRI over antenatal US in diagnosing both common and unusual posterior fossa abnormalities. The value of MRI is even more pertinent in developing countries where third trimester unbooked antenatal presentations are common; one should have a low threshold for performing MRI at referral institutions in such patients.

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