

Diagnosis of Chiari III malformation by second trimester fetal MRI with postnatal MRI and CT correlation

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Abstract We report a case of Chiari III malformation diagnosed by fetal MRI. Ultrasound (US) performed at a gestational age of 18 weeks demonstrated a posterior skull base cyst. Repeat US at 19 weeks demonstrated neural tissue in the cyst, consistent with an encephalocele. MR imaging at 23 weeks confirmed the presence of an occipital encephalocele, demonstrated additional bony defect in the upper cervical spine, and identified abnormal morphology and position of the brainstem consistent with the diagnosis of Chiari III. Postnatal MRI and CT confirmed the fetal MRI findings and demonstrate the utility of fetal MRI in the early evaluation of sonographically detected posterior fossa abnormalities.

Keywords Chiari III · Fetal MRI · Occipital encephalocele

Introduction

In 1891, Hans Chiari first described the Chiari III malformation. The infants were noted to have herniation of the cerebellum through a cervical spina bifida with associated caudal displacement of the medulla [1]. The

definition has since been broadened to include low occipital encephaloceles. It is a very rare disorder, and the largest case series consists of only nine patients [2]. In a study of 312 children with Chiari complex, only two had Chiari III malformations [3]. Although prenatal sonography can detect cephaloceles, it may not reliably characterize the contents of the cyst or identify associated CNS anomalies. Prenatal MRI often complements sonography because of its ability to evaluate the developing parenchyma, cortex and posterior fossa structures [4, 5]. We report a case where early prenatal diagnosis of Chiari III malformation was identified using fetal MRI.

Case report

A 20-year-old gravida 1 para 0 female was referred for antenatal MRI after prenatal ultrasound (US) at 18 weeks estimated gestational age (EGA), which revealed a cystic mass in the occipital region with no underlying calvarium. The biparietal diameter was 35 mm, consistent with an EGA of 17 weeks and 1 day. A more detailed US at 19 weeks demonstrated neural tissue extending into the 2.0×2.7-cm cyst consistent with an encephalocele. No other abnormalities were identified; however, the posterior fossa was not well seen (Fig. 1). The patient was given the diagnosis of an occipital encephalocele and was referred for fetal MRI.

Fetal MRI was performed at 23 weeks EGA on a 1.5-T MR scanner (GE Healthcare, Milwaukee, Wis) with an eight-channel, torso phased-array coil. Axial, coronal, and sagittal single-shot fast spin-echo T2-weighted images of the fetal brain were obtained (TR 6,666 ms, TE 91 ms, 3-mm slice thickness, 0-mm skip). A defect in the inferior aspect of the occipital bone as well as the upper cervical spine was identified (Fig. 2). There was an associated 3.1×6.1×4.4-cm

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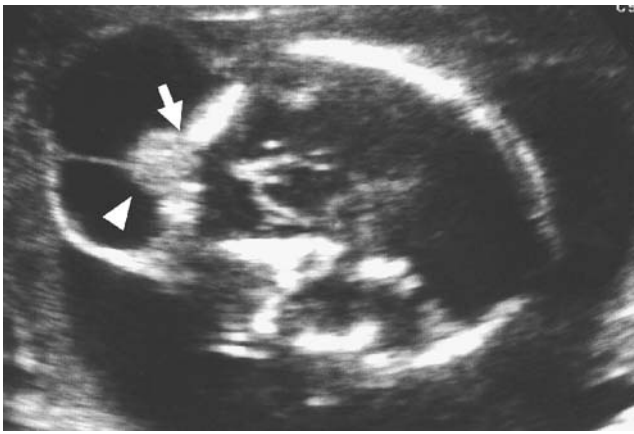
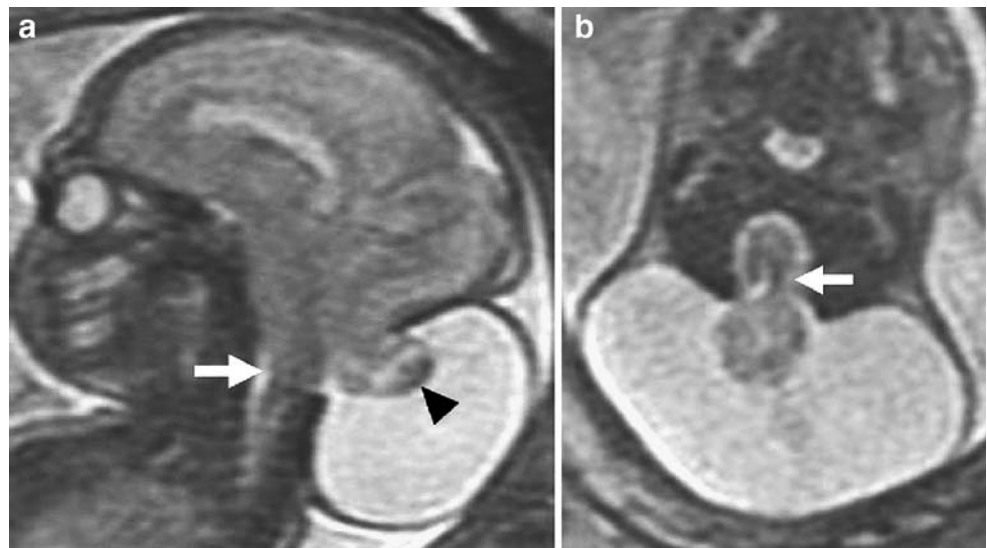


Fig. 1 Sonographic image of the fetal head at 19 weeks' EGA, demonstrates a cystic structure arising from the occipital region. A defect in the calvarium is identified (*arrow*) through which neural tissue is noted to protrude (*arrowhead*)

cystic structure with signal characteristics similar to cerebrospinal fluid. Neural tissue, which was contiguous with the cerebellum and inferior occipital lobes, protruded through the defect into the cystic structure consistent with an occipital and high cervical encephalocele. Within the herniated neural tissue was a central ovoid of T2 prolongation consistent with a displaced fourth ventricle. Additional smaller areas of T2 prolongation were consistent with areas of encephalomalacia in the herniated cerebellar tissue. The posterior fossa was small, with inferior displacement and cervicomedullary kinking of the brainstem. The brainstem was elongated and extended to, but not into, the posterior bony defect. There was associated diffuse effacement of the subarachnoid spaces and abnormal angulation of the Sylvian fissures and parieto-occipital sulci. Due to the distortion of the corpus callosum from the inferior displacement of the posterior aspect of the cerebrum, the splenium of the corpus callosum was not visualized, suggesting possible hypogenesis.

Fig. 2 Sagittal single-shot fast spin-echo T2-W image performed at 23 weeks' EGA demonstrates a low occipital and high cervical bony defect through which neural tissue (*arrowhead*) herniates into a cystic structure consistent with an encephalocele. **a** Small posterior fossa, effacement of the subarachnoid spaces, and inferior displacement of the brainstem with cervicomedullary kinking (*arrow*) is consistent with a Chiari III malformation. **b** Axial single-shot fast spin-echo T2-W image demonstrates abnormal morphology of the brainstem, which extends to the margin of the bony defect (*arrow*)

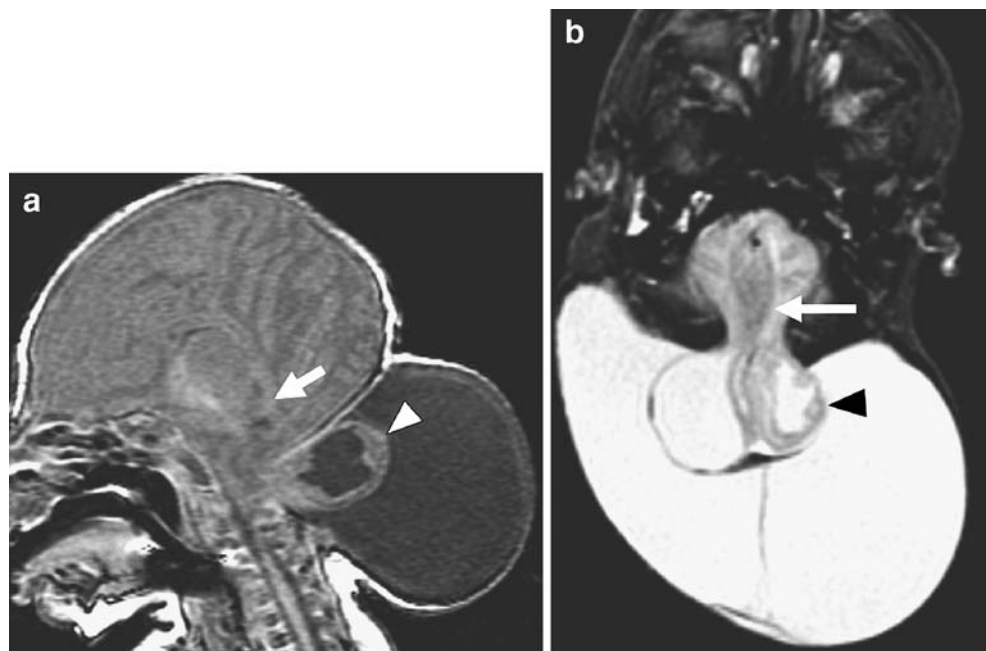


The third ventricle was inferiorly positioned and the right and left lateral ventricles measured 12 mm and 5 mm in atrial diameter, respectively. Based on the MRI findings, the diagnosis of Chiari III malformation was made, and the patient was counseled about the neurodevelopmental prognosis. The parents were also counseled to deliver at a tertiary hospital to assure the availability of a pediatric neurosurgeon and neonatal intensive care unit for the anticipated postnatal surgical repair.

The infant was born at 38 weeks EGA by cesarean section, which is standard care in our institution for infants with encephaloceles. The infant weighed 3,390 g, and Apgar scores were 7 and 8. The baby was vigorous at delivery and received supplemental oxygen for 5 min. The head circumference was 33 cm. The encephalocele was covered with normal skin and measured 14 cm in the transverse dimension and 8 cm in the craniocaudal dimension. There was normal tone and movement of all four extremities.

MRI performed on the 1st day of life confirmed the presence of a large occipital and high cervical encephalocele (Fig. 3). There was increased cystic encephalomalacia involving the herniated neural tissue compared with the antenatal MRI, with no residual cerebellum. A portion of the occipital and temporal lobes herniated inferiorly into the posterior fossa and extended laterally and ventrally to the brainstem. The brainstem was inferiorly displaced and extended to the ventral aspect of the cephalocele, unchanged in appearance from the fetal MRI. The corpus callosum was present, but thinned posteriorly accounting for the difficulty in visualization of the splenium on the prenatal MRI due to the limited resolution. The sulci were also abnormal in morphology, with abnormal angulation and depth of sulci, but without frank cortical malformation, which is an appearance frequently observed in children

Fig. 3 MRI on the first day of life. **a** Sagittal 3-D spoiled gradient recalled echo T1-W image confirms the fetal MRI findings of a Chiari III malformation. The occipital lobes extend into the encephalocele with increased cystic encephalomalacia (*arrow*) compared to the fetal MRI. The corpus callosum is intact (*arrowhead*). **b** Axial spin-echo T2-W image confirms the abnormal morphology and position of the brainstem (*arrow*) as well as increased cystic encephalomalacia of the herniated occipital lobes (*arrowhead*). No normal cerebellum was identified



with Chiari II malformation. The ventricles were normal in size. The dural venous sinuses were anomalous. The sagittal sinus divided into three dural venous sinus structures that extended inferiorly. In retrospect, an anomalous venous sinus was present in the midline on the prenatal MRI, but the inherent limited resolution of the images made it difficult to detect. A bony defect was seen involving the inferior aspect of the occipital bone and posterior elements of the C1 vertebral body. CT imaging with 3-D reformations confirmed the defect involving the occipital bone and posterior aspect of the C1 arch (Fig. 4). The posterior arches of C2 were slightly splayed apart. The posterior arches of C3, C4, C5 and C6 were all unfused. The lamboid, sagittal, and coronal sutures were diastatic.

The encephalocele was resected on day of life 10. The dural defect measured 3 cm at the time of surgery. Pathology revealed disorganized reactive glioneuronal neuropil most consistent with an occipital encephalocele, along with fragments of cerebellar tissue in the encephalocele. At 2 months of age, the baby developed hydrocephalus requiring placement of a ventriculoperitoneal shunt. The baby is currently 3 months old and has increased tone in all extremities, seizure disorder, episodes of central apnea, and requires supplemental nutrition via a gastrostomy tube.

Discussion

The Chiari III malformation is a rare anomaly defined by the presence of a low occipital and/or high cervical encephalocele. Some features such as a small posterior fossa, cervicomedullary kinking, herniation of cerebellar

tonsils into the foramen magnum, tectal beaking, corpus callosum dysgenesis, and hydrocephalus are shared with Chiari II malformations. Agenesis of the posterior elements of upper cervical vertebrae is a frequent finding. This is

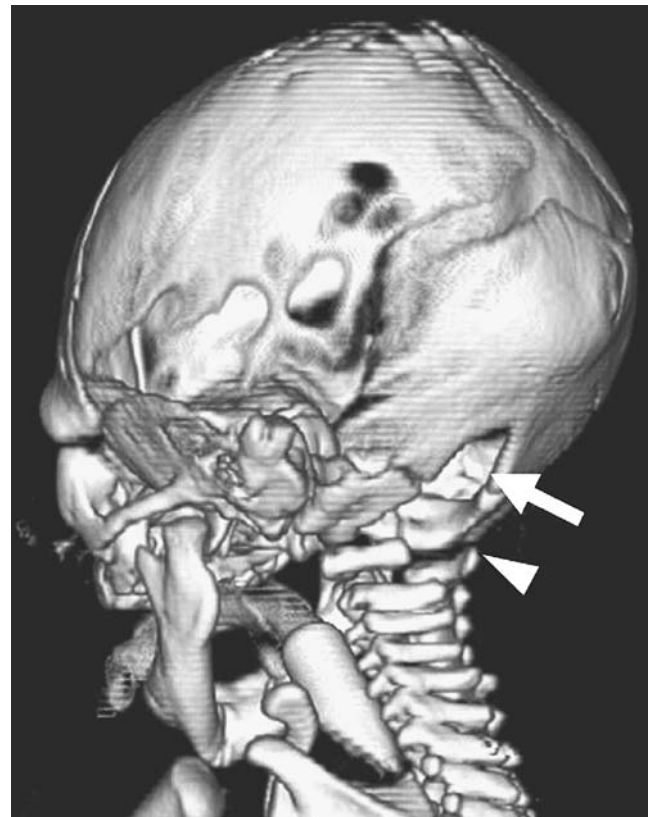


Fig. 4 The 3-D reformation from an axial CT scan demonstrates the defect in the occipital bone (*arrow*) and posterior aspect of the C1 vertebral body (*arrowhead*)

most common at the C1 level and can be seen in up to 70% of cases of Chiari III [2, 6]. In our patient, bony anomalies of the occipital bone and upper cervical spine were detected by fetal MRI and characterized further by postnatal CT imaging with 3-D reformations.

Although US is the modality of choice for evaluation of fetal anomalies it can be limited by maternal body habitus, fetal position, oligohydramnios, and ossification of bony structures. Recent studies have demonstrated that prenatal MRI is helpful in evaluating specific brain abnormalities, especially those involving the posterior fossa [4, 5]. In our case, both US and fetal MRI clearly identified neural tissue within the cephalocele. However, the antenatal MRI allowed better characterization of this tissue by demonstrating direct contiguity with the cerebellum and inferior occipital lobes. Moreover, the fetal MRI also identified involvement of the brainstem and upper cervical spine, and led to the prenatal diagnosis of Chiari III malformation.

The information provided by the prenatal MRI was critical for prenatal counseling regarding the neurodevelopmental prognosis, since the parents were counseled about the outcome of patients with Chiari III malformation, rather than the outcome of isolated encephaloceles. More specifically, the patient was counseled regarding the frequent incidence of hydrocephalus, developmental delay, cranial nerve deficits, respiratory insufficiency, and hypertonica in children with Chiari III malformations [3, 6]. The fetal MRI also influenced obstetrical decisions about the mode and location of delivery. In addition, the antenatal MRI provided the pediatric neurosurgeon with critical information needed to initially plan the postnatal surgical procedure and to more accurately discuss the risks and prognosis of the procedure with the parents prior to delivery.

Prenatal MR imaging of Chiari III in a 34-week fetus has been reported [7]. In our case, antenatal MRI was performed much earlier, during the second trimester, and many of the abnormalities seen postnatally could already be visualized at 23 weeks' gestation. In addition, the 2nd trimester fetal MRI was able to demonstrate associated abnormalities, including the abnormal inferior position of the brainstem and extension to the bony defect, which is important since identification of brainstem involvement is critical to postnatal management

[2]. Indeed, this child has episodes of central apnea as well as diffusely increased tone.

It is interesting that the extent of encephalomalacia in the herniated occipital and cerebellar tissue was increased on the postnatal MRI as compared to the prenatal MRI. This most likely resulted from interval increased destruction of the herniated tissue, either due to venous and/or arterial compromise, or possibly direct compression of the tissue, which has been described in the setting of Chiari II malformations [8]. The fetal MRI, however, was limited in its evaluation of the posterior corpus callosum and of the dural venous sinuses, in part due to the distortion from the encephalocele. In retrospect, with attention to the flow voids on the fetal MRI, we were able to discern the anomalous venous drainage, which is frequent in children with Chiari III and which is important in the planning of the neurosurgical repair of the defect [3, 7].

In conclusion, fetal MRI is a useful adjunct to prenatal sonography when a posterior skull base cephalocele is identified. Knowledge of the appearance of Chiari III malformation and its associated abnormalities during the 2nd trimester is important because accurate diagnosis may affect patient counseling and clinical management.

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