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Case Report

Imaging in the presence of meroanencephaly

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ABSTRACT

Meroanencephaly occurs when there is an incomplete open median calvarial defect. This condition, which is in the spectrum of anencephaly, results in ectopic brain without skin covering and a normal foramen magnum. We present a rare case of a female fetus with meroanencephaly referred to our institution at 24 weeks and imaged with both prenatal ultrasound and MRI, demonstrating an open neural tube defect in the high parietal area and lack of visualization of the supratentorial ventricular system. Postnatal the child survived and went on to require antibiotic therapy and closure of the defect without cerebral spinal fluid diversion but demonstrates severe permanent neurologic deficits.

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Introduction

Neural tube defects are a common referral for imaging, particularly in the fetus to provide counseling with regard to outcome and direct decisions with regard to intrauterine and postnatal care. Cranial neural tube defects can be open or closed, with anencephaly being the most common form of open neural tube defect resulting in failed closure of the rostral end of the neural tube [1]. Encephaloceles are the most reported closed form with the defect described by calvarial location [2].

Meroanencephaly is a rare open cranial defect in the spectrum of anencephaly. The calvarial defect occurs in the superior midline with deficiency of overlying skin. Given incom-

plete coverage of the median cerebrum, brain disorganization occurs, resulting in lack of normal cerebral landmarks, including absence of a supratentorial ventricular system. It is important to consider this entity as it may mimic an encephalocele in the parieto-occipital area. In this report, we present a description of prenatal and postnatal imaging findings and postnatal outcome in the presence of meroanencephaly.

Case Report

This case report was approved by our Institutional Research Board (IRB) and signed informed consent to use de-identified medical information for research purposes was obtained from

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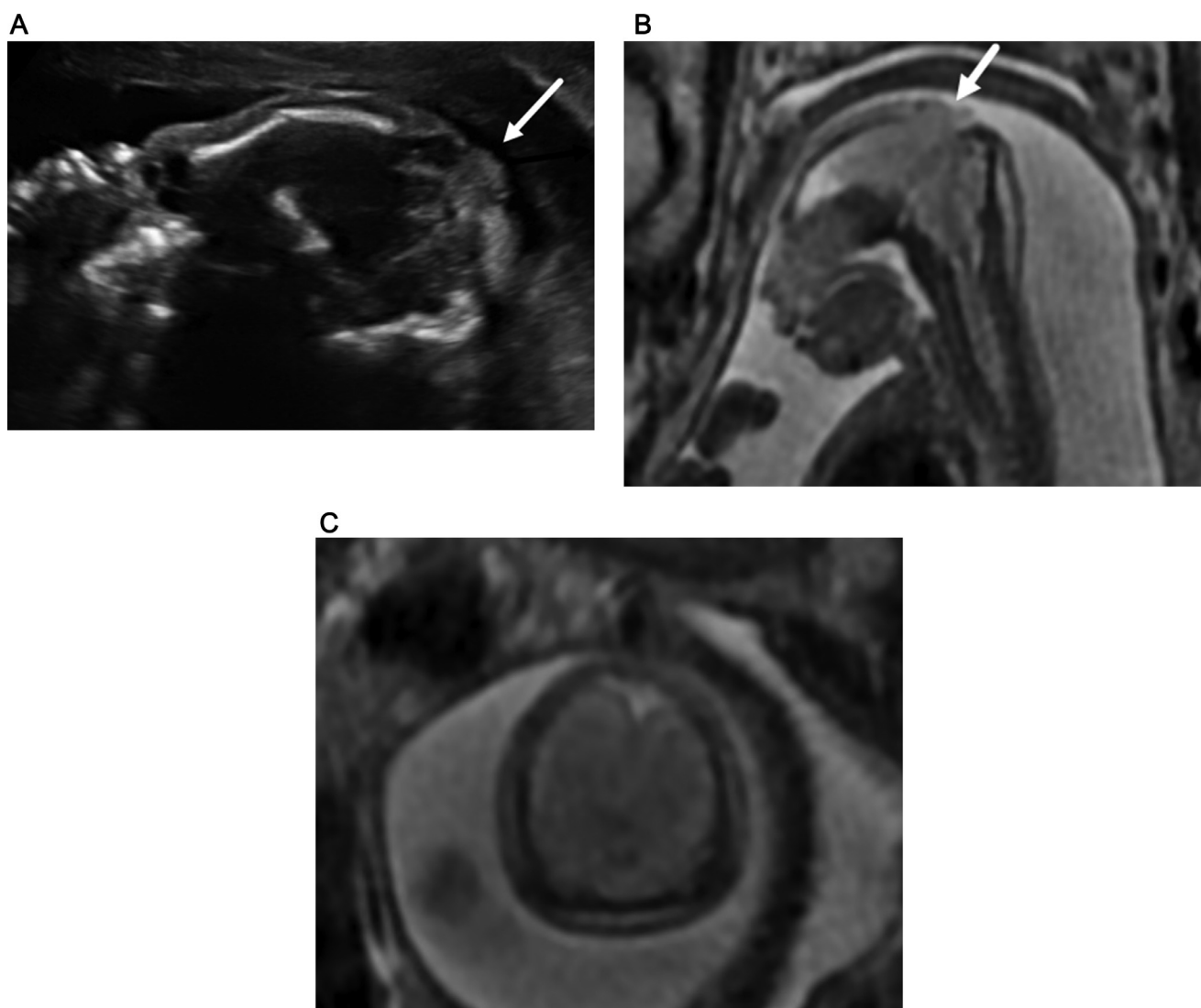


Fig. 1 – Meroanencephaly with imaging in a female fetus at 24 weeks and 6 days. A: Sagittal ultrasound demonstrates a defect in the calvarium with herniation of dysmorphic tissue through the defect (arrow). No skin covering suggested. B: Sagittal T2-SSFSE image from fetal MRI demonstrates similar findings of midline calvarial defect, herniation of disorganized tissue (arrow) and no apparent skin covering. C: Axial T2-SSFSE image from fetal MRI shows lack of lateral ventricles, germinal matrix and sulcation.

the patients at initial evaluation and retained within the electronic medical records system.

A 23-year-old G1PO woman was referred to our center at 24 weeks and 6 days for concern for fetal microcephaly. Ultrasound and fetal MRI were performed on presentation. Ultrasound brain biparietal diameter and head circumference were less than third percentile for gestational age. A midline calvarial defect was noted with associated herniation of meninges and dysmorphic brain tissue (Fig. 1A). There was lack of normal cerebral landmarks, and the supratentorial ventricular system could not be identified. The posterior fossa was small and transcerebellar diameter difficult to measure due to distorted anatomy. The nuchal fold was normal and no extracranial anomalies were present. In fact, the face was normal, and amniotic fluid volume was normal. Fetal MRI con-

firmed a high parietal midline calvarial defect in the area of the posterior fontanelle with herniation of small disorganized brain and meninges without a defined sac/skin covering (Fig. 1B and C). The supratentorial brain was small and lacked normal anatomy, including absent lateral ventricles, germinal matrix, brain lamination pattern and supratentorial midline anatomy. The posterior fossa was small with cerebellum and brainstem distorted toward the calvarial defect. The family was counseled that the prognosis for the child was poor and that the child would likely not survive given the open nature of the defect.

The child was born near their home, out of state at 39 6/7 weeks by C-section and did not pass away at birth. Therefore, the family returned to our center at day 9 of life for further evaluation. The child was noted to have a 3 cm midline

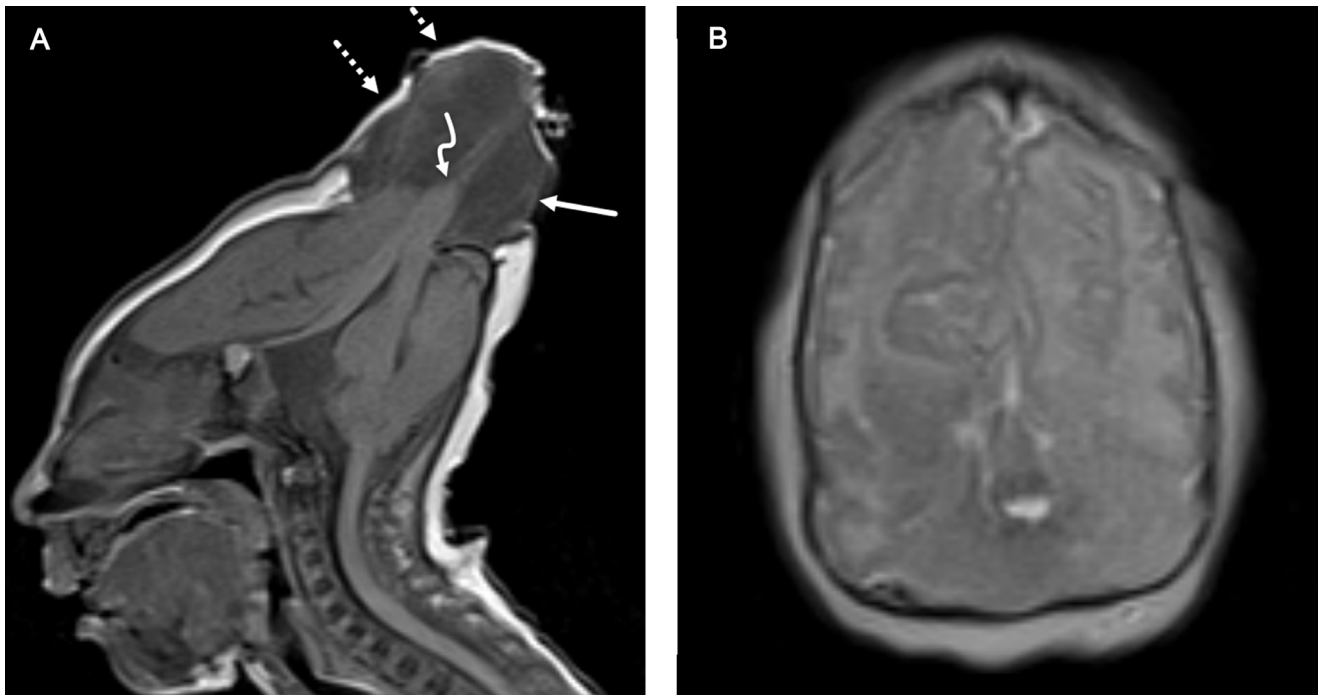


Fig. 2 – Postnatal imaging of meroanencephaly of same patient from [Figure 1](#) at 10 days of life. **A:** Sagittal T1 image shows median calvarial defect in area of posterior fontanelle with herniation of disorganized tissue and CSF (solid arrow). High signal material outlining defect (dotted arrows) were dressings over the open tissue. The brainstem is stretched and pulled into the defect with the curved arrow representing the midbrain. The posterior fossa is distorted but there is normal development of the cerebellum and lower brainstem. **B:** Axial T2-FSE MRI again demonstrates lack of ventricular system with complete effacement of the extra-axial CSF spaces and disorganized supratentorial brain.

defect in the area of the posterior fontanelle with no skin or meningeal covering and exposed malformed brain protruding approximately 2 cm from the defect. Head circumference was <1% based on WHO standards. The child was placed on prophylactic antibiotics and imaging, both MRI and CT venogram, was performed. Postnatal MRI confirmed the defect in the midline. The supratentorial brain was small and lacked presence of a normal cerebral ventricular system ([Fig. 2A](#) and [B](#)). A CT venogram showed sagittal and transverse sinus in the defect. Unfortunately, the child developed infection within the open defect that was treated with broad spectrum antibiotics. Seizures began at 1 month of age. At 2 months of age, closure of the defect with cranioplasty was performed. The child did not require ventricular shunting and is currently 3 years of age. She is nonambulatory, unable to sit independently, non-verbal, severe global delays and difficult to control epilepsy.

Discussion

Congenital absence of the calvarium leads to exposure of the brain tissue to amniotic fluid and physical trauma, which results in varying degrees of brain destruction. The anencephaly phenotype falls along a spectrum based on how much of the calvarium is congenitally absent. While holoacrania results in classic anencephaly, merocrania is a milder form of anen-

cephaly in which the defect is open but the calvarial defect is small [1]. Meroanencephaly occurs in 3.4% of cases referred for the suspected diagnosis of anencephaly, [3] and there are very few cases of meroanencephaly in the literature [4–6]. A pathology paper describes merocrania in a case of a child that had clear demarcation of the cerebrum, cerebellum and brainstem but with amorphous and disorganized brain [4]. A publication of three cases of meroanencephaly reported that two of the fetuses were aborted and not followed to delivery and the third was stillborn [5]. A single case reports survival of a child postnatal [6].

In the presence of disorganized brain and a neural tube defect, the differential should include anencephaly, acrania, meroanencephaly, craniorachischisis, encephalocele, and amniotic band syndrome. Identifying a midline calvarial defect with variable brain, cerebrospinal fluid and venous sinuses contained in a skin covered sac is important in diagnosing an encephalocele. Amniotic band calvarial defects may be open but are usually off midline and often associated with other organ deformities or amputations [7]. Anencephaly, acrania and craniorachischisis are the most severe open neural tube defects and lethal [8–10]. Acrania is a mesodermal defect with absence of calvarium, and craniorachischisis is due to closure defects of both the brain and spinal column.

The importance in correctly diagnosing this malformation and discriminating from an encephalocele is that the patient is born with an open cranial defect that can get infected.



Fig. 3 – Prenatal and Postnatal Meroanencephaly. A: Sagittal T2-SSFP image from prenatal MRI at 29 weeks and 6 days in a female fetus showing midline parietal calvarial defect with herniation of dysmorphic tissue without skin covering (arrow). Notice lack of midline anatomy or visualized ventricles. **B:** Postnatal T2-FSE MRI of the same patient on day of life 1 showing similar findings with arrow again denoting open neural tube defect.

Therefore, closure of the defect promptly would be appropriate and if that is not possible, administration of prophylactic antibiotics may be warranted.

Patient counseling in the setting of meroanencephaly may differ from anencephaly. This child did not demise intrauterine or at birth as is typical for anencephaly [8]. Therefore, unlike anencephaly, this milder variant is not necessarily lethal. This is supported in the literature [6] and by a second case that we encountered at our institution with similar findings (Fig. 3 A and B) who is alive at 10 years of age. This child with meroanencephaly is G-tube dependent with significant developmental delay, spastic quadriplegic, epilepsy and cortical visual impairment. Therefore neurologic sequela is severe in comparison to encephaloceles where outcome is dependent on location, herniation of brain parenchyma, presence of microcephaly and anomalies of the intracranial contents [2]. In addition, children with encephaloceles typically require ventricular shunting after closure, and this second child with meroanencephaly has not required ventricular shunting given absent supratentorial ventricular system.

In summary, in the presence of a midline superior calvarial defect in a fetus, the differential should include meroanencephaly, especially when supratentorial anatomy is distorted and supratentorial ventricular system cannot be defined. These findings are helpful in discriminating from an encephalocele. Unlike anencephaly, those with meroanencephaly can survive; however, in our experience, neurologic outcome is poor.

REFERENCES

- [1] Goldstein RB, Filly RA. Prenatal diagnosis of anencephaly: spectrum of sonographic appearances and distinction from the amniotic band syndrome. *Am J Radiol* 1998;151:154–550.
- [2] Thompson DNP. Postnatal management and outcome for neural tube defects including spina bifida and encephaloceles. *Prenatal Diagnosis* 2009;29:412–19.
- [3] Pelizzari E, Melendez Valdez C, Picetti JS, Campos da Cunha A, et al. Characteristics of fetuses evaluated due to suspected anencephaly: a population-based cohort study in southern Brazil. *Sao Paulo Med J* 2015;133(2):101–8.
- [4] Melnick M, Myrianthopoulos NC. Studies in neural Tube Defects II. Pathologic findings in a prospectively collected series of anencephalics. *Am J Med Genet* 1987;26:797–810.
- [5] Isada NB, Qureshi F, Jacques SM, et al. Meroanencephaly: Pathology and prenatal diagnosis. *Fetal Diagnosis Ther* 1993;8:423–8.
- [6] Khan IA, Firdaus U, Ali SM, Asghar I. Newborn with meroanencephaly: Surviving all odds. *J Pediatric Neurosci* 2016;11(3):228–9.
- [7] Sentilhes L, Verspyck E, Patrier S, et al. Amniotic band syndrome: pathogenesis, prenatal diagnosis and neonatal management. *J Gynecol Obstet Biol Reprod* 2003;32:693–704.
- [8] Blencow H, Kancherla V, Moorthis S, et al. Estimates of global and regional prevalence of neural tube defects for 2015: a systemic analysis. *Ann N Y Acad Sci*. 2015;1414:31–46.
- [9] Avagliano L, Massa V, George TM, et al. Overview on neural tube defects: from development to physical characteristics. *Birth Defects Res* 2018. doi:10.1002/bdr2.1380.
- [10] Weissman A, Diukman R, Auslender R. Fetal acrania: five new cases and review of the literature. *J Clin Ultrasound* 1997;25:511–14.