AMSER Case of the Month December 2023

A female infant born at 38 weeks 1 day with a cranial defect and exposed brain tissue

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Patient Presentation

- **HPI:** A female infant born at 38 weeks 1 day to a 35-year-old G3P3003 mother via planned C-section due to frank breech position. Patient weighed 2160g at birth. APGAR scores at 1 minute and 5 minutes were 8 and 9, respectively.
- **Physical Exam:** Infant was alert and active with eyes open. Proptosis noted. Anterior fontanelle flat, sunken, and soft. Open median parietooccipital cranial defect with exposed brain tissue. No overlying dural coverings, with a small amount of CSF leakage.
- **Medical History:** Third trimester fetal anatomy ultrasound showed the following: Abnormal, small brain parenchyma with partial calvarium and small residual brain tissue. Posterior placenta with normal amniotic fluid volume. Fetus varies between cephalic and footling breech presentation.
    - Repeat ultrasound nine days later indicated frank breech position.

Pertinent Labs

- Prenatal:
  - Maternal Serum Alpha-Fetoprotein (AFP): 421.8 (nl at 15-20 weeks: 10-150 ng/ml).
- Postnatal:
  - Newborn metabolic screen within normal limits. Elevated bilirubin of 6.9 (nl: 0.2-1.2 mg/dL).

What Imaging Should We Order?

ACR Appropriateness Criteria

Variant 1: **Neonatal seizures, age 0 to 29 days. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
MRI head without IV contrast	Usually Appropriate	O
US head	May Be Appropriate	O
MRI head without and with IV contrast	May Be Appropriate	O
CT head without IV contrast	May Be Appropriate	☢☢☢
CT head with IV contrast	Usually Not Appropriate	☢☢☢
CT head without and with IV contrast	Usually Not Appropriate	☢☢☢☢
HMPAO SPECT or SPECT/CT brain	Usually Not Appropriate	☢☢☢☢
FDG-PET/CT brain	Usually Not Appropriate	☢☢☢☢

This imaging modality was ordered. Variant 1 was used since physical exam findings raised concern for congenital brain structural abnormalities

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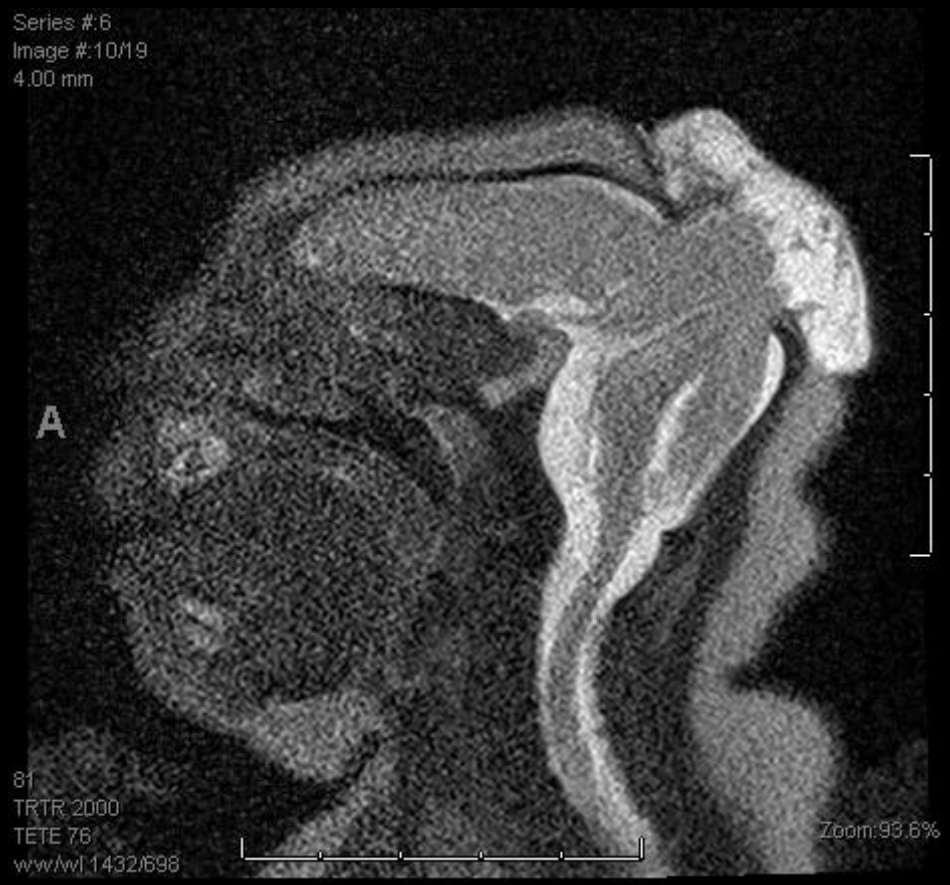
Findings (Unlabeled)

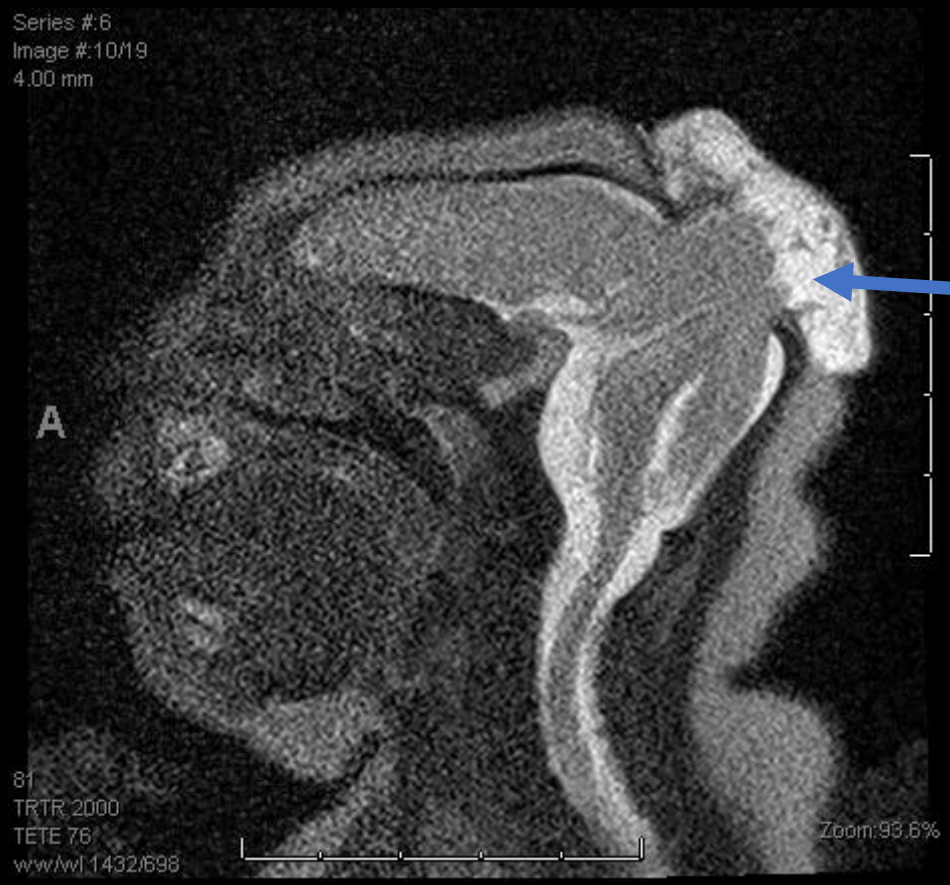
- Rapid unenhanced, unsedated brain MRI (single-shot turbo-spin echo sequence)



Sagittal

Findings

- Rapid unenhanced, unsedated brain MRI (single-shot turbo-spin echo sequence)

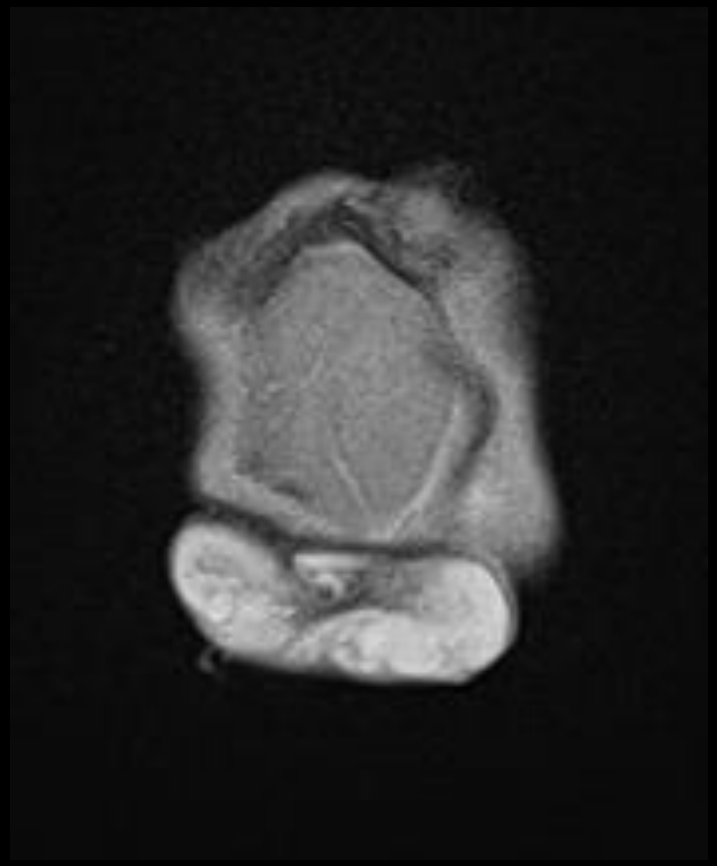


Midline cranial defect measuring up to 2 cm, with disorganized brain protruding through

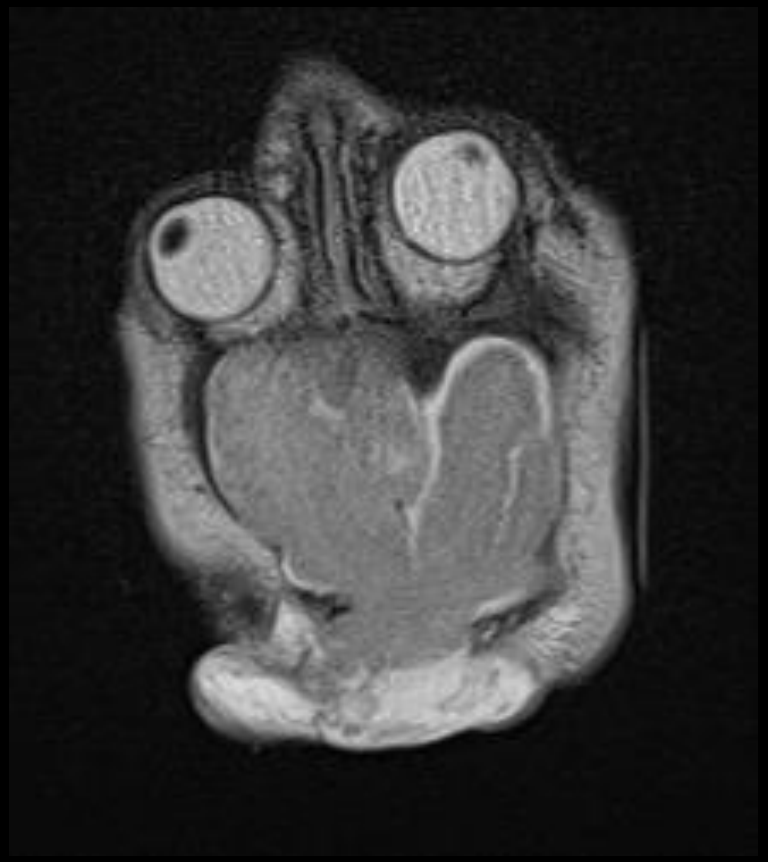
Sagittal

Findings (Unlabeled)

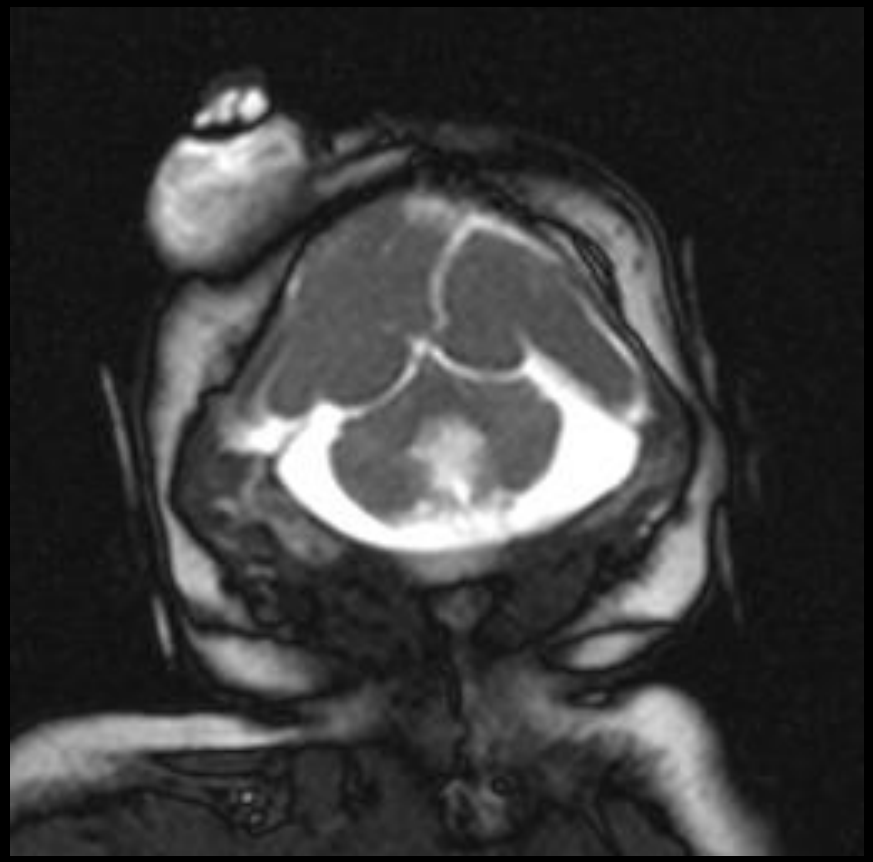
- MRI brain shunt series without IV contrast



Axial T2



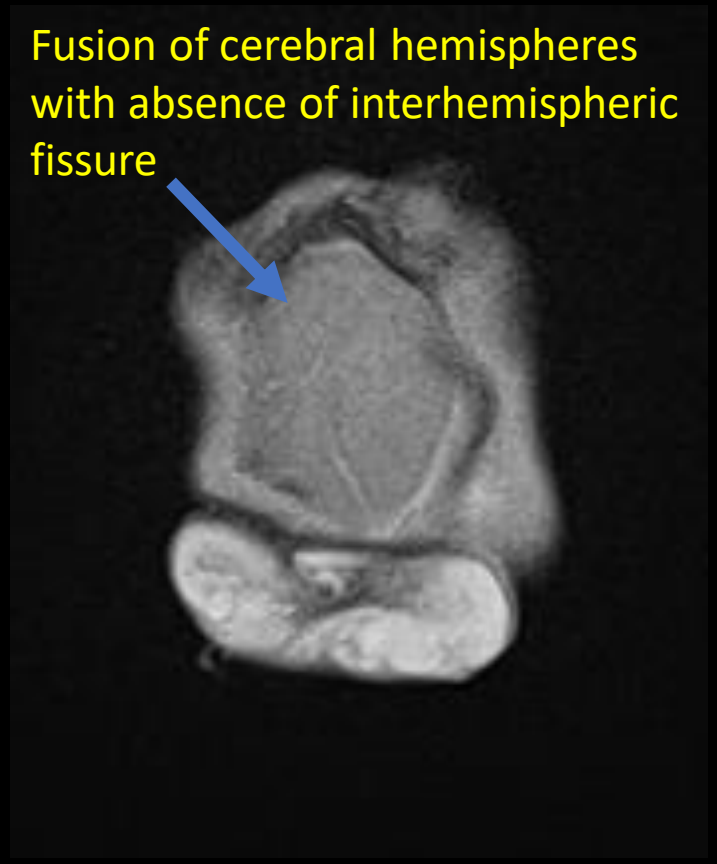
Axial T2



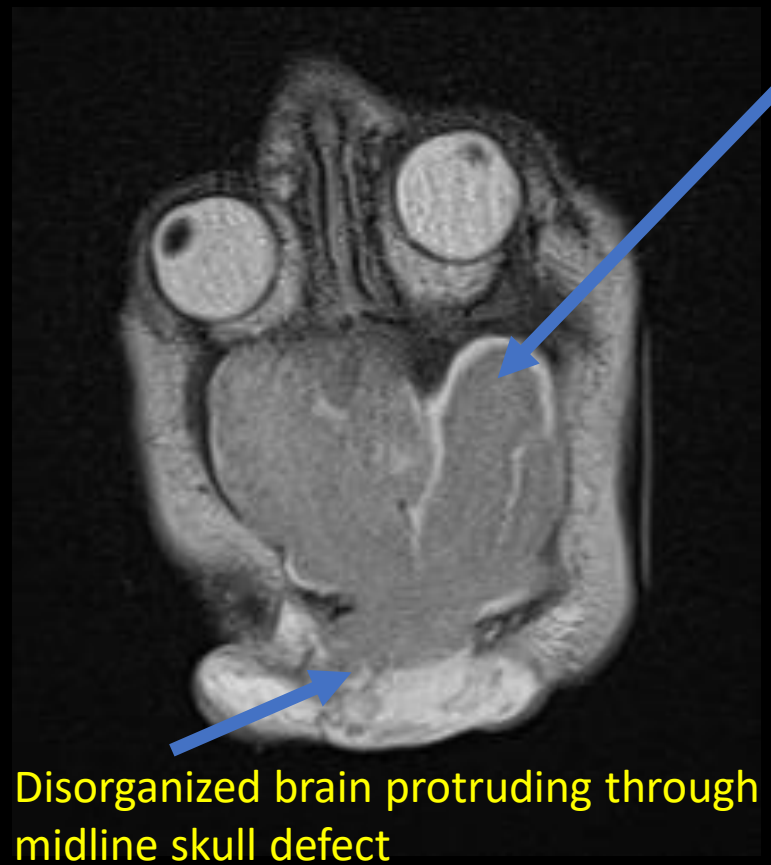
Coronal TrueFISP

Findings

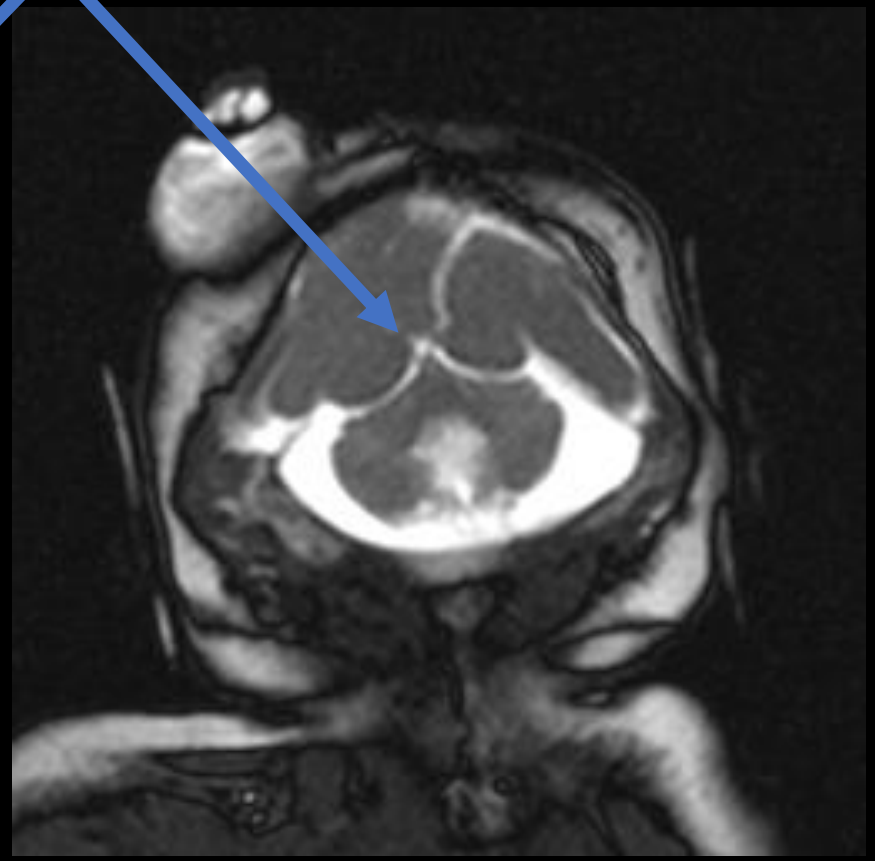
- MRI brain shunt series without IV contrast



Axial T2



Axial T2



Coronal TrueFISP

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Final Dx:

Meroanencephaly

Case Discussion

- Patient admitted to NICU for 3 days and discharged home with comfort care measures. Wound care (with Mepilex Border + Exufiber Ag) of defect discussed with parents.
- Medications at Discharge:
  - Lorazepam (0.2 mg every 4 hours as needed)
  - Morphine (0.2 mg every 2 hours as needed)

Case Discussion

- Meroanencephaly is characterized by partial malformation of the cranium and rudimentary brain tissue. There is also an open median cranial defect through which the area cerebrovasculosa protrudes. Area cerebrovasculosa is comprised of abnormal spongy, vascular tissue and glial tissue.[1]

- Falls within the acrania-exencephaly-anencephaly (AEA) sequence, in which incomplete absence of the calvarium results in progressive destruction of dysplastic brain.[2]

- Anencephaly, the end of the sequence in which there is no forebrain/midbrain/hindbrain, is estimated to have an incidence of 1 in every 4,600 births, with a female predominance of 3:1.[2,3]

Case Discussion

- Defects across the AEA sequence have a multifactorial etiology including both genetic and environmental factors. Inadequate dietary folate consumption before conception is the most relevant.[2]

- Elevated maternal serum alpha-fetoprotein (AFP) and low estriol levels are highly sensitive though diagnosis is confirmed using prenatal ultrasound.
  - **Ultrasound Findings:** Widened cranial pole, asymmetric disorganized brain tissue, absence of hyperechogenic calvarial ossifications, decreased head-to-trunk ratio, echogenic amniotic fluid.[2]

- MRI may be used when ultrasound imaging is limited.

- There is no medical treatment with efforts focused on palliative care.

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References:

1. Isada, N. B., Qureshi, F., Jacques, S. M., Holzgreve, W., Tout, M. J., Johnson, M. P., & Evans, M. I. (2009). Meroanencephaly: Pathology and Prenatal Diagnosis. *Fetal Diagnosis and Therapy*, *8*(6), 423–428. https://doi.org/10.1159/000263862

2. Kline-Fath, B. M., Bulas, D., & Lee, W. (2020). In *Fundamental and advanced fetal imaging ultrasound and MRI* (2nd ed., pp. 627–629). Wolters Kluwer.

3. CDC. (2023, June 28). *Facts about Anencephaly | CDC*. Centers for Disease Control and Prevention. https://www.cdc.gov/ncbddd/birthdefects/anencephaly.html

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