AMSER Case of the Month February 2025

36 y.o. G6P2032 female presenting for further evaluation of multiple anomalies seen on 17-week ultrasound

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Patient Presentation

- HPI: 36-year-old female with gestational diabetes and history of several miscarriages and ectopic pregnancy presents at 28 weeks gestation for evaluation of anomalies found on 17-week ultrasound. Patient reports normal fetal movement and denies contractions, leakage of fluid, or vaginal bleeding.
- Past Medical History: G6P2032, including 2 cesarean sections (1 elective, 1 emergent); hyperlipidemia
- Medications: Baby aspirin; prenatal multivitamin; magnesium citrate; atorvastatin
- Social History: Heavy cigarette use

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Noninvasive Prenatal Testing (NIPT)

- Fetal sex: male
- Negative for trisomies or microdeletion syndromes

What Imaging Should We Order?

ACR Appropriateness Criteria

Variant 4: **Second and third trimester screening for abnormal finding on ultrasound: major anomalies. Next imaging study.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal detailed scan	Usually Appropriate	O
MRI fetal without IV contrast	Usually Appropriate	O
US echocardiography fetal	Usually Appropriate	O
US pregnant uterus transabdominal follow-up	Usually Appropriate	O
MRI fetal without and with IV contrast	Usually Not Appropriate	O

This imaging modality was ordered by the physician

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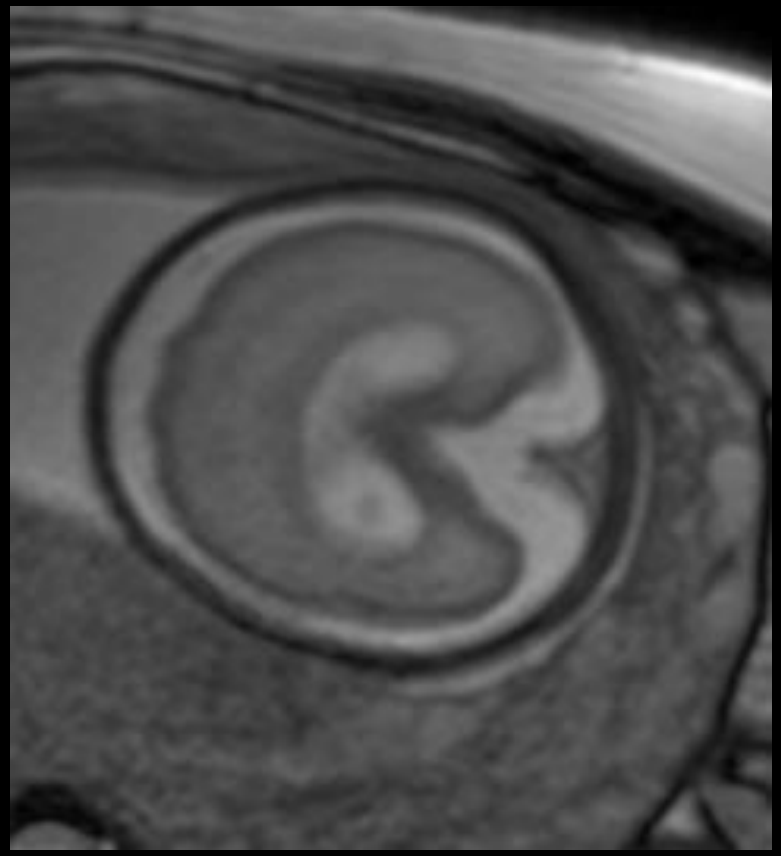
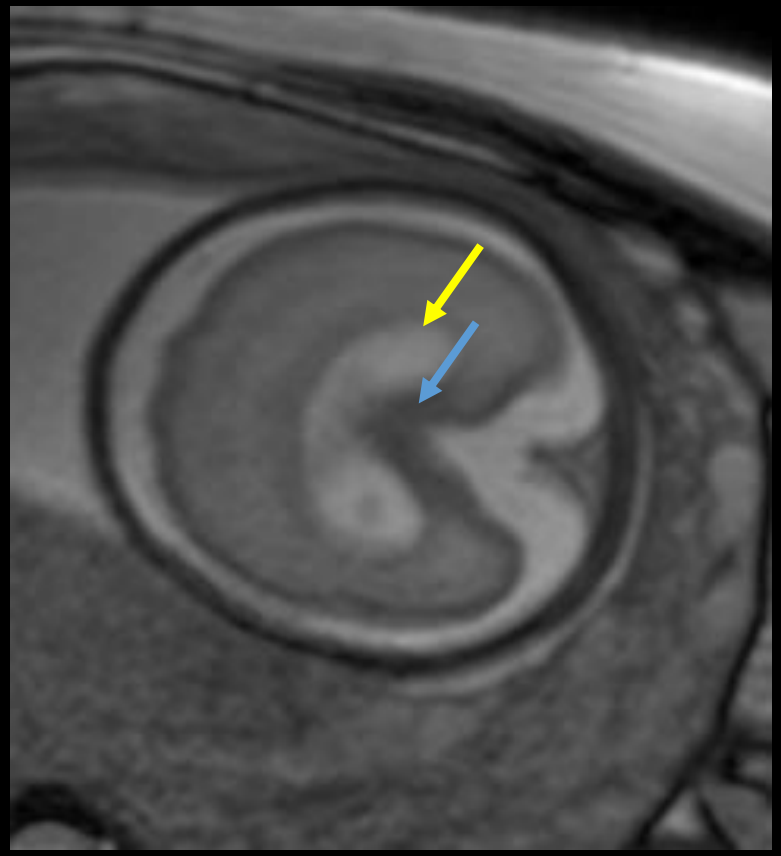
Findings (unlabeled)

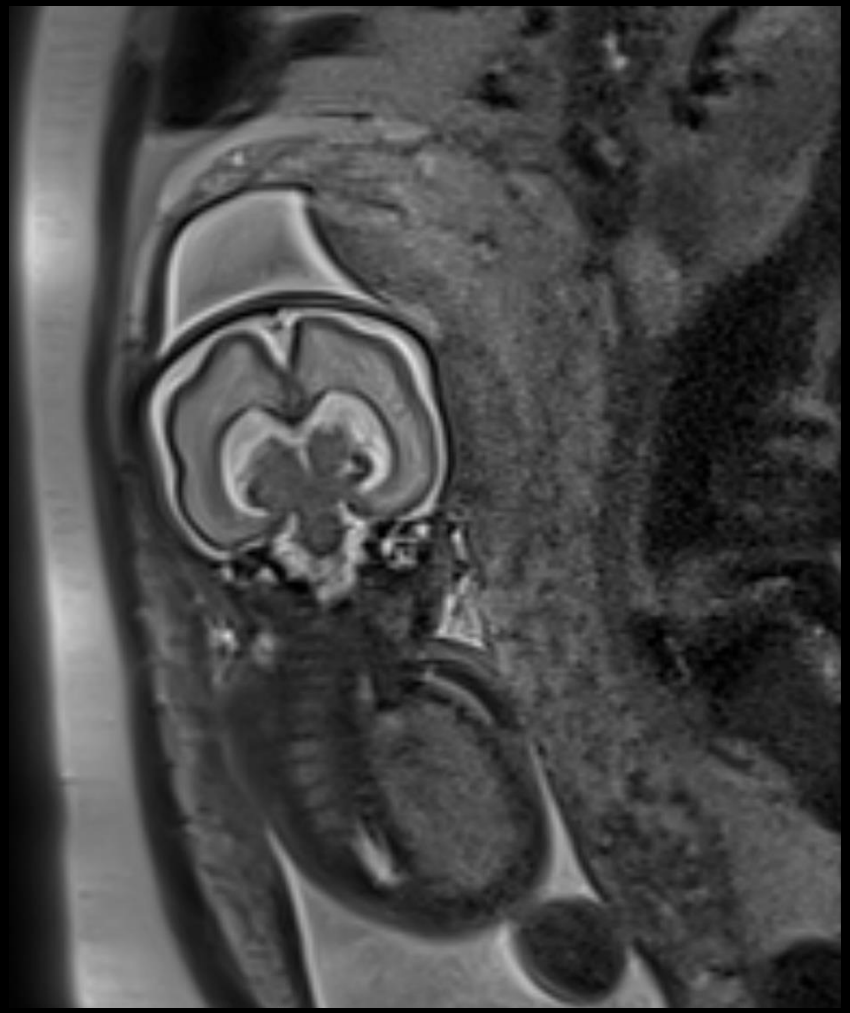


Findings (labeled)



Axial T2 image shows a monoventricle (arrow), with absence of the septum pellucidum and fusion of the anterior cerebral hemispheres (arrow)

Findings (unlabeled)



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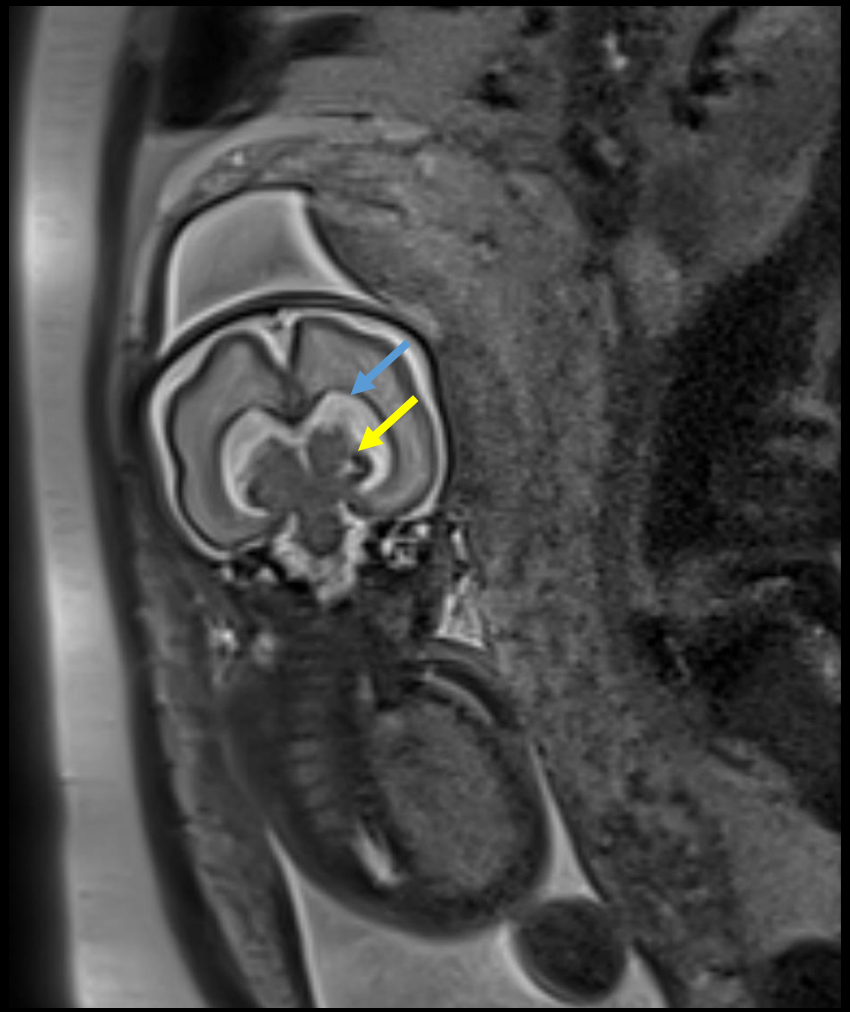
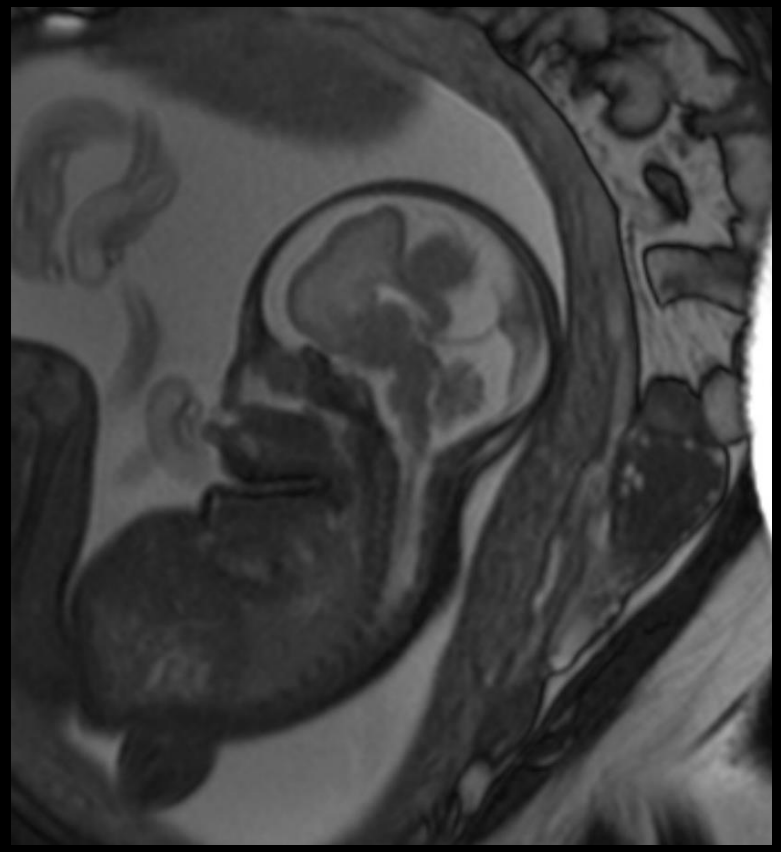
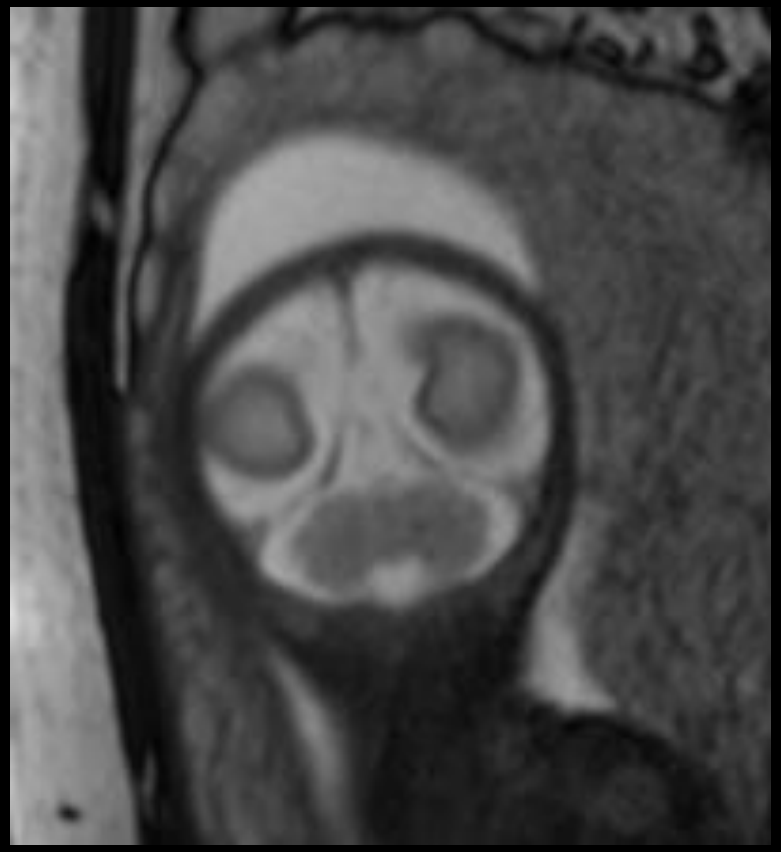
Findings (labeled)



Coronal T2 image shows incomplete separation of the thalami (arrow), with a monoventricle (arrow)

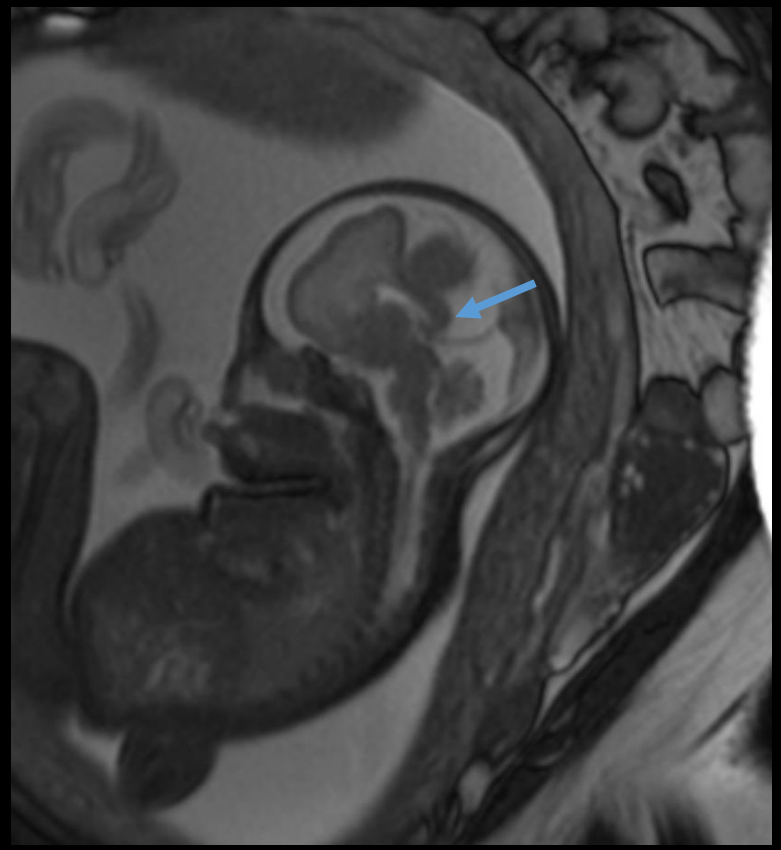
Findings (unlabeled)

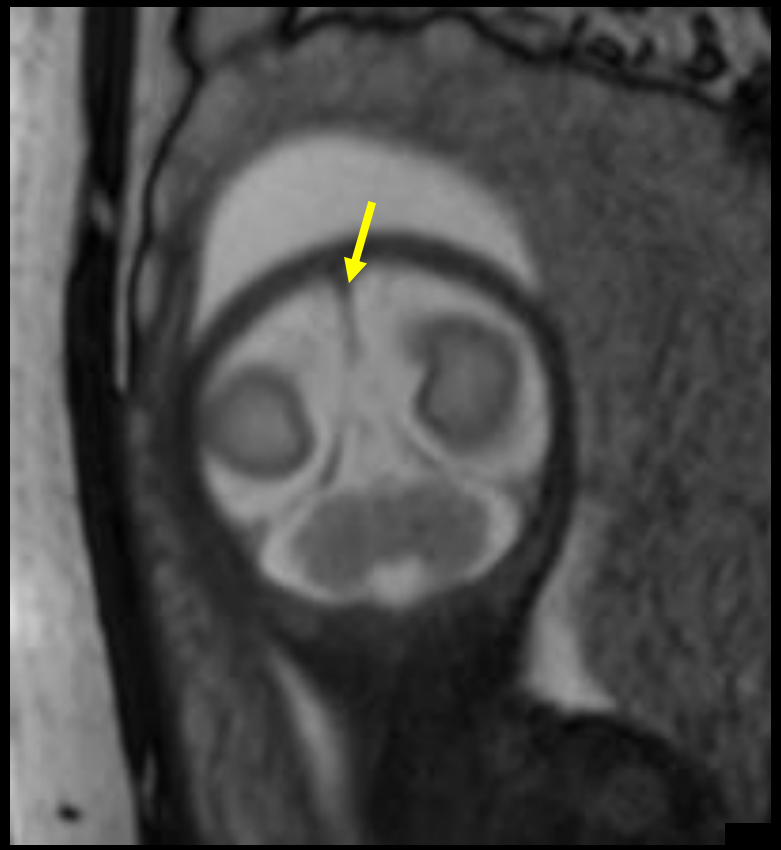




Findings (labeled)

Dysplastic appearance of the posterior corpus callosum (arrow)





Presence of the posterior falx cerebri, with separation of the posterior cerebral hemispheres

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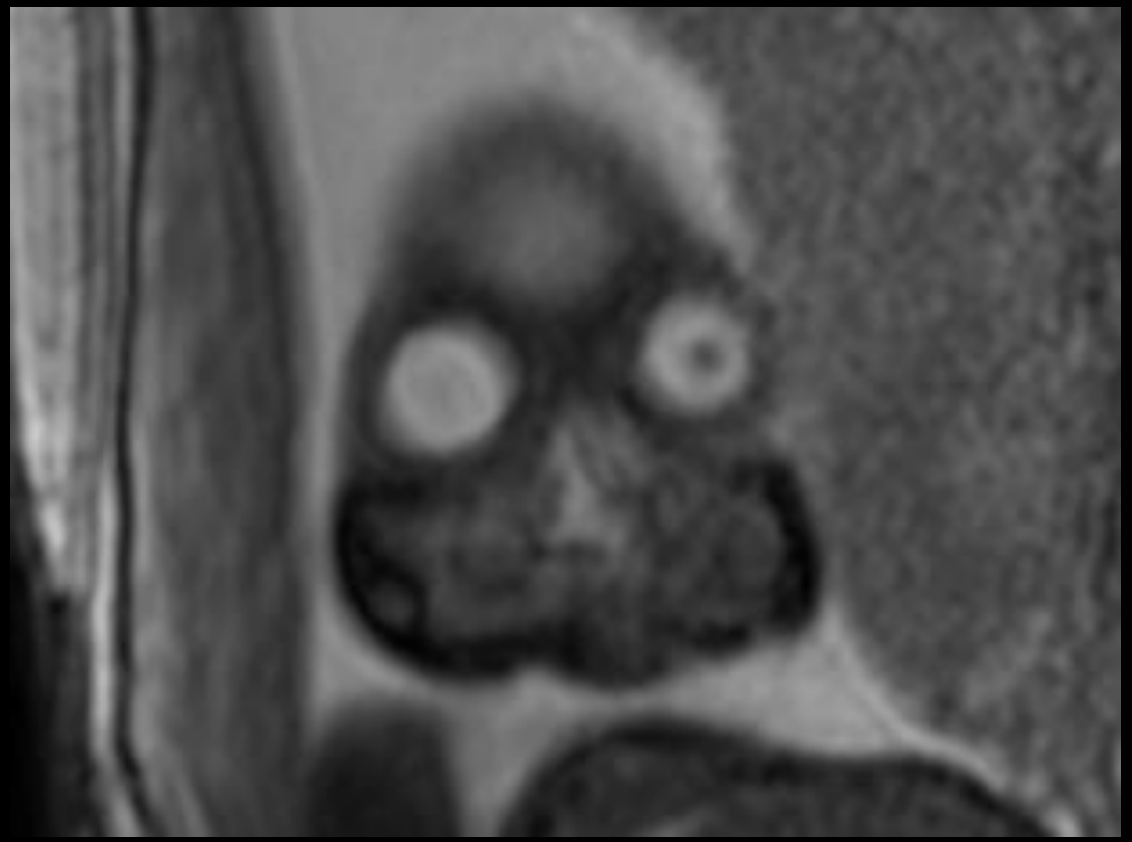
Findings (unlabeled)



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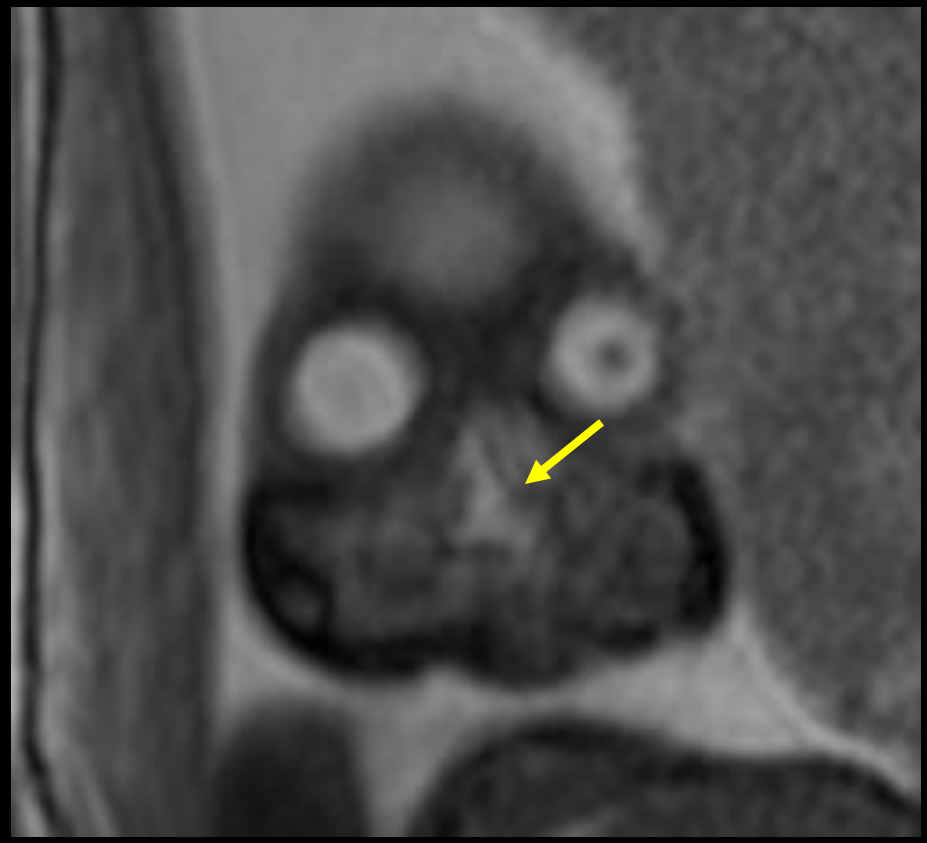
Findings (labeled)



Cleft lip and palate (arrow)

Final Dx:

Semilobar Holoprosencephaly

Case Discussion

- Background
  - Holoprosencephaly (HPE) is a complex congenital disorder characterized by a spectrum of incomplete separation of the cerebral hemispheres due to anomalous signaling mechanisms between the neural crest and neural ectoderm.[3]
  - The three classic forms of HPE include lobar, semilobar, and alobar, which differ by their degree of forebrain cleavage; alobar is the most severe.[2]
  - In general, cerebral cleavage progresses from posterior to anterior.
  - In semilobar HPE, premature cessation of cleavage results in partial falx cerebri formation and fusion of the anterior cerebral forebrain.[1]
- Epidemiology
  - The prevalence of HPE ranges from 0.48 to 1.70 per 100,000 births.[2]
  - In the United States, HPE is slightly more prevalent among African American, Hispanic, and Pakistani communities, which is suspected to reflect reduced prenatal diagnosis.[3]
  - Polysomic abnormalities account for 25-50% of cases (i.e., trisomy 1, 3, 13, 18, 19) and can be found incidentally in monogenic syndromes (i.e., Smith-Lemli-Opitz syndrome).[2]

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Case Discussion

- Clinical Presentation
  - HPE confers a poor prognosis, with a 70-80% mortality rate during the first year of life.[3]
  - Disease manifestations include developmental delay, feeding difficulties, epilepsy, instability of temperature, heart rate, and respiration.
  - Associated endocrine abnormalities include diabetes insipidus, adrenal hypoplasia, hypogonadism, thyroid hypoplasia, and growth hormone deficiency.[1]
  - Patients may present with facial anomalies, such as cleft lip and palate, cyclopia, proboscis, and hypotelorism.[1]
- Imaging Findings
  - HPE is commonly detected by ultrasound and confirmed with fetal MRI.
  - MRI best demonstrates a horseshoe-shaped monoventricle, with absence of the septum pellucidum.[3]
- Management
  - Discussion of a birth plan with parents is valuable prenatally.
  - Multidisciplinary management is essential, including anticonvulsive therapy, physical and occupational therapies, and endocrine disorder testing.[1]

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References:

1. Dubourg C, Bendavid C, Pasquier L, et al. Holoprosencephaly. *Orphanet J Rare Dis.* 2007 Feb 2;2:8. doi: 10.1186/1750-1172-2-8.
2. Gomez GD, Corrêa DG, Trapp B, et al. Holoprosencephaly spectrum: an up-to-date overview of classification, genetics and neuroimaging. *Jpn J Radiol.* 2025 Jan;43(1):13-31. doi: 10.1007/s11604-024-01655-8.
3. Ramakrishnan S, Das JM. Holoprosencephaly. Holoprosencephaly. 2024 Jun 7. In: *StatPearls* [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan–.

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