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Subtle anomalies of fetal CNS

Roya Sohaey MD, Professor, Diagnostic Radiology

Date: May 2014

Goal

- Practical approach to detectable brain anomalies that are subtle and important.
  - Diagnosis is suggestible on routine axial views

<u>Anatomic approach</u>

- Anterior brain
- Lateral ventricles
- Parenchyma
- Posterior fossa

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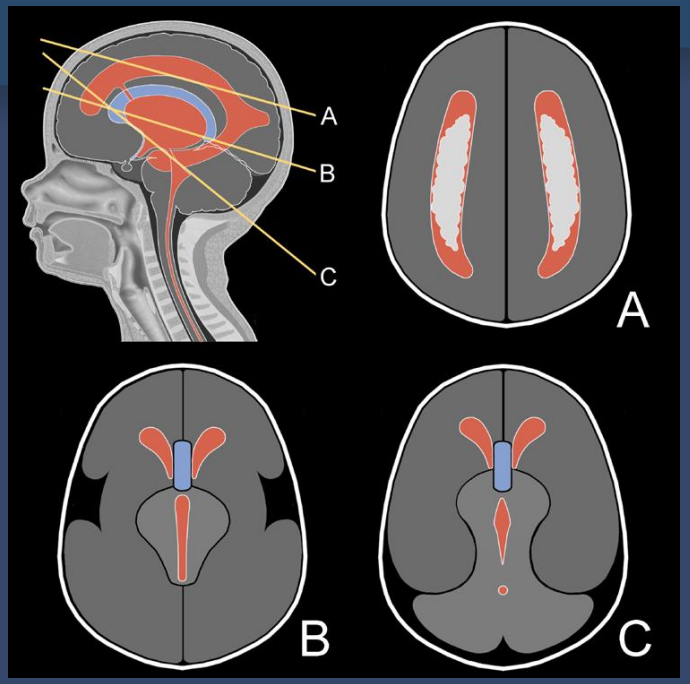
Routine Axial Views: AIUM, ACR guidelines

- 3 routine views
  - Lateral ventricles
  - BPD/HC level of thalamus
  - Posterior fossa
- Structures you should see from these views
  - Midline falx
  - Cavum Septi Pellucidi
  - Choroid plexus
  - Lateral ventricles
  - Cerebellum, including vermis
  - Nuchal fold
- Total yield: 3 views, 2 biometry measurements, at least 7 anatomic structures!
- Consider additional views to clear anatomy or problem solve



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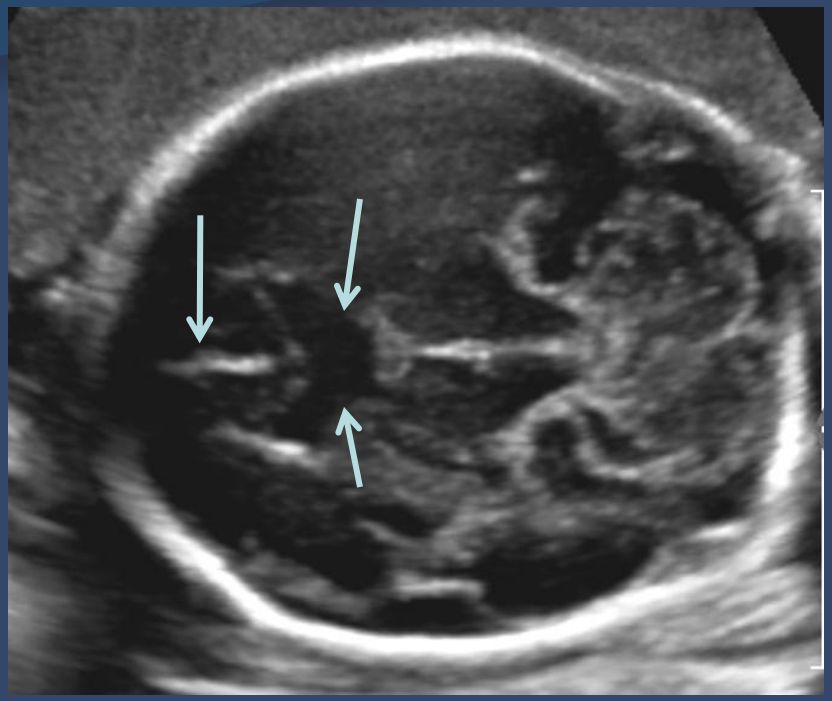
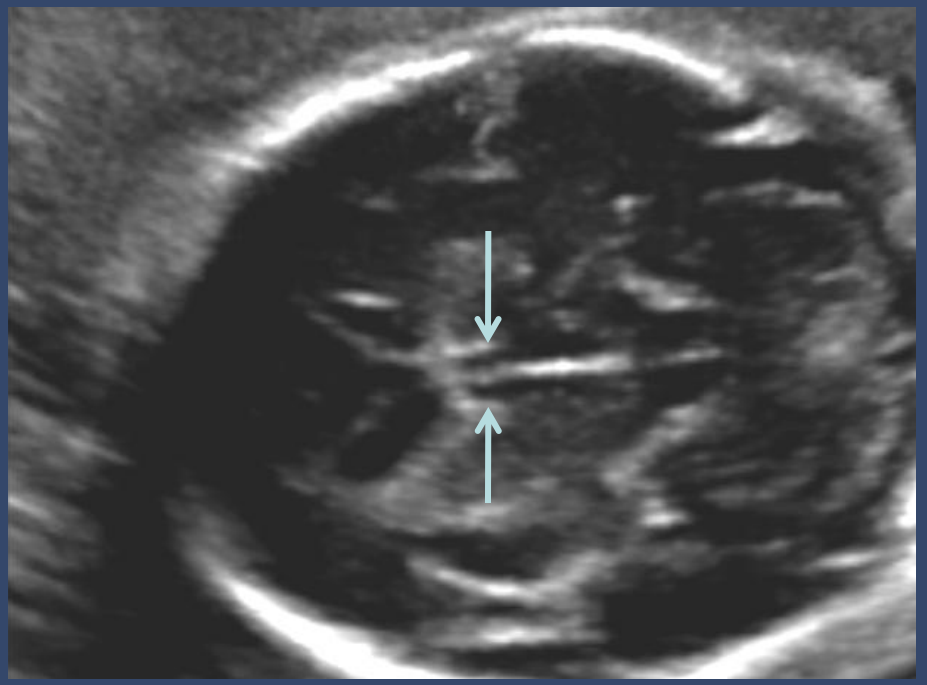
Case: Is anterior anatomy normal? Falx? CSP?





- Falx: Yes
- CSP: NO!
- TP: Don’t confuse the normal paired fornices are not CSP
  - Caudal (inferior) to CSP

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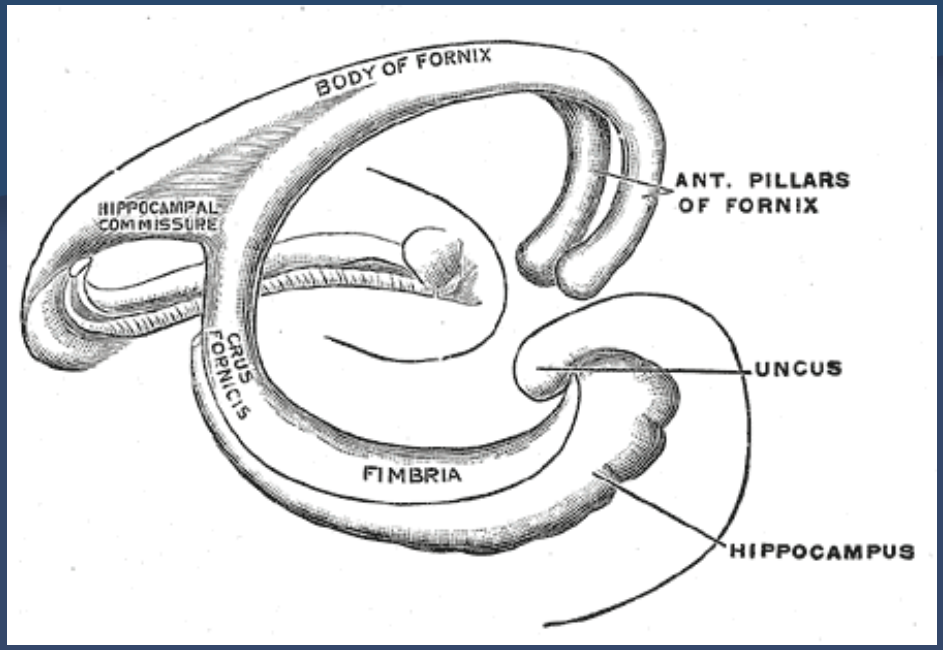
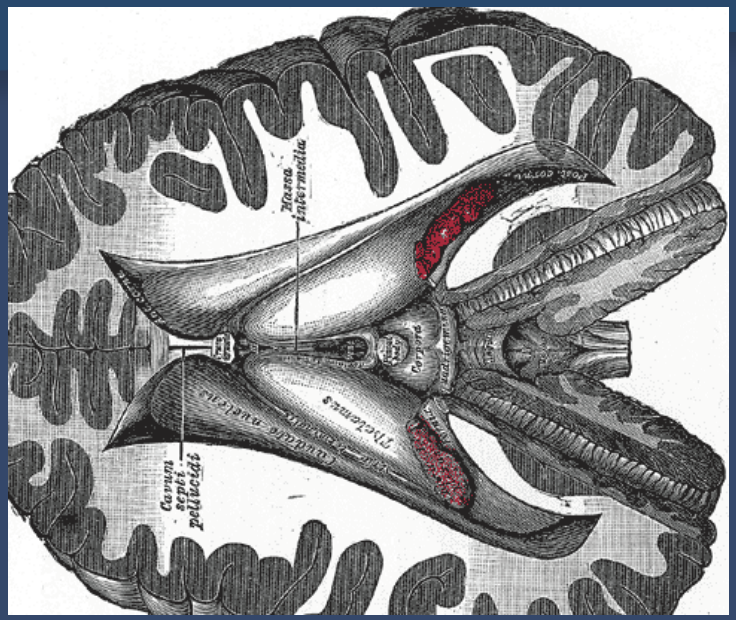
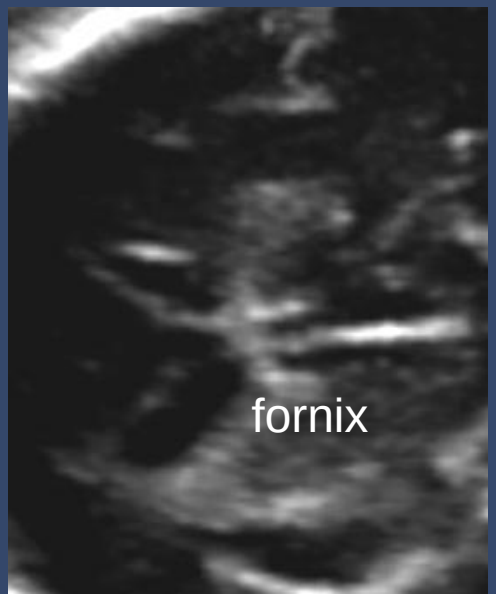
Cavum Septi Pellucidum vs Fornix







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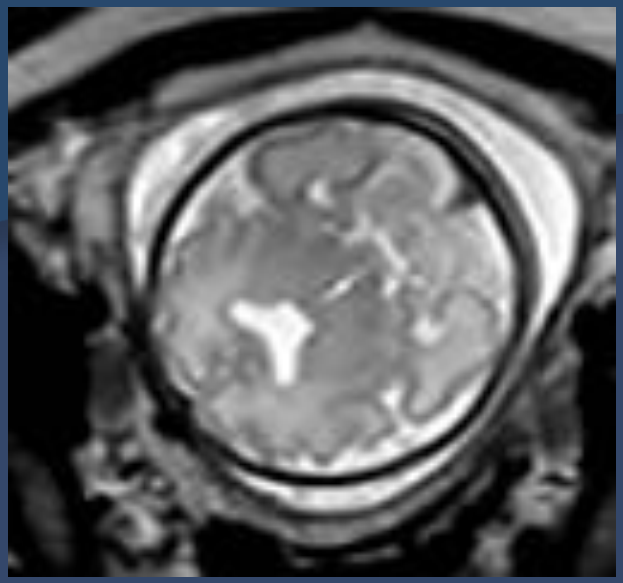
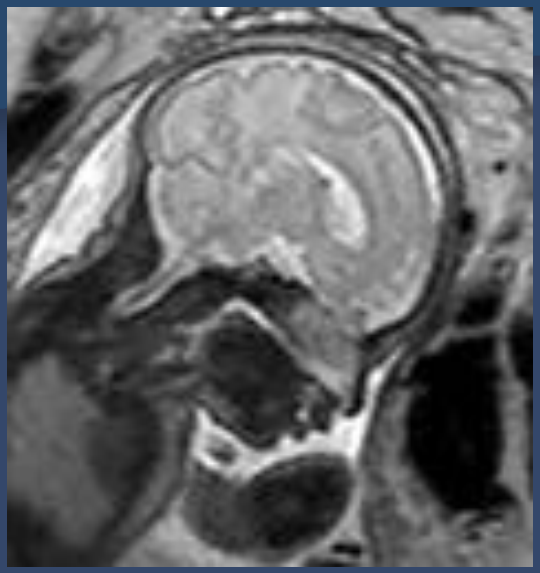
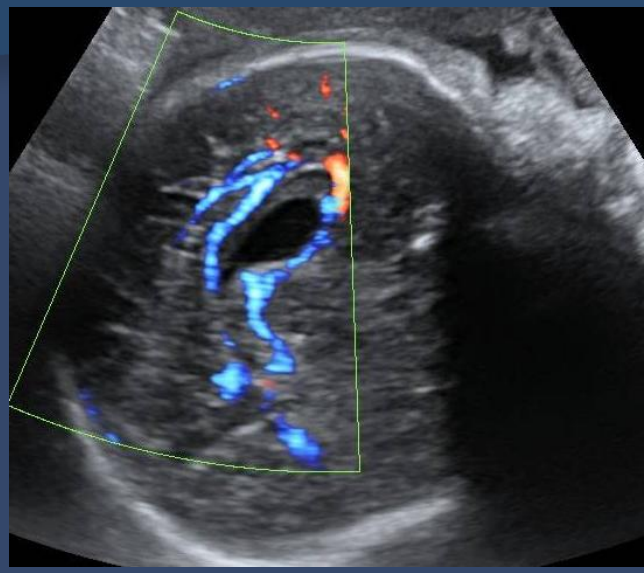
Case: Absent CSP, additional fetal imaging







- Absent Cavum: MR to look for additional anomalies (not for confirmation)
- Normal Corpus Callosum
- Normal brain parenchyma
- Isolated absent CSP???
- “your baby will be fine”???

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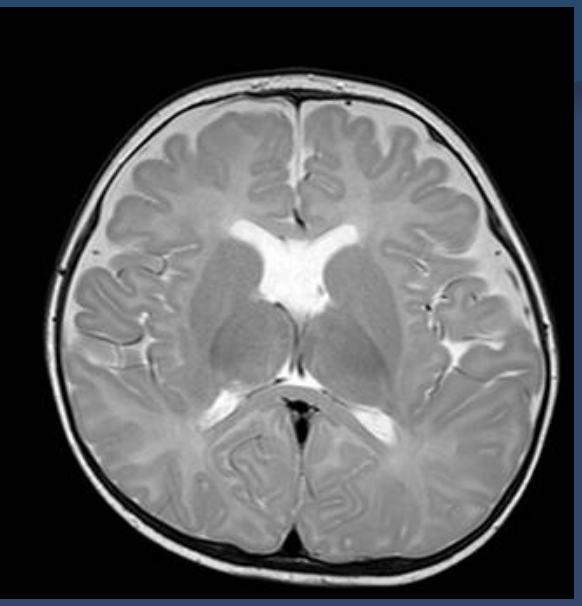
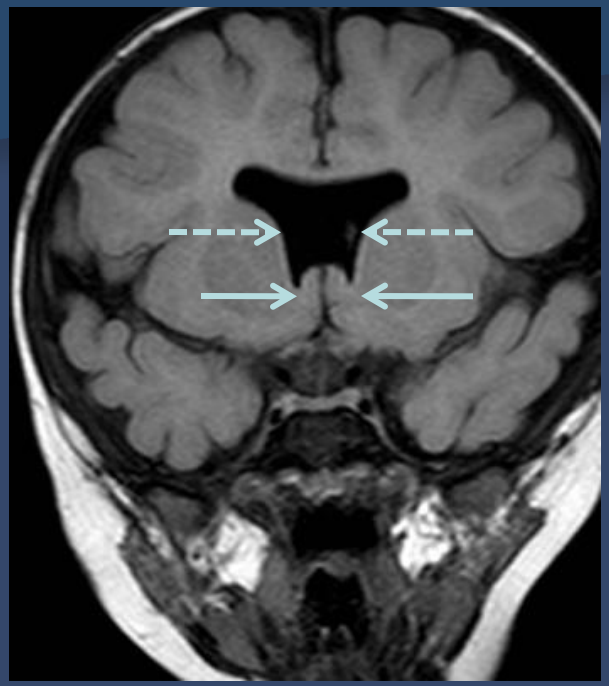
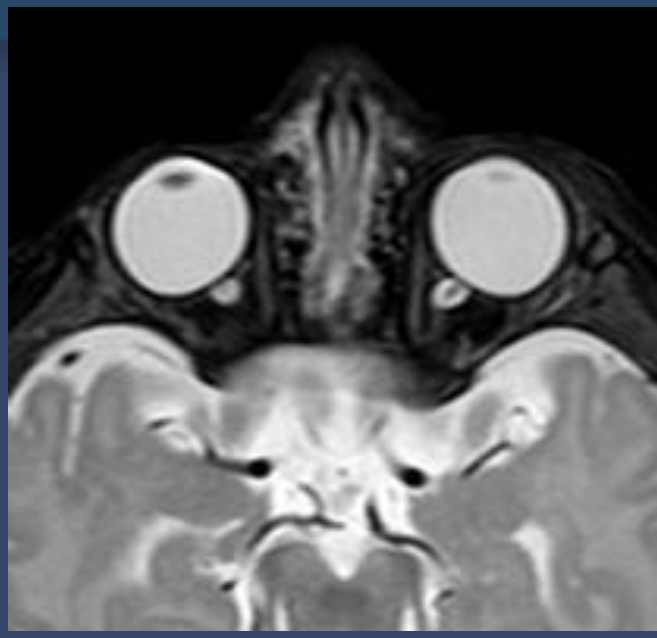
Case: Neonatal MR







- Final diagnosis: Septo-optic Dysplasia
- Child with severe nystagmus to light and essentially blind.
- Absent CSP, hypoplastic optic nerves and chiasm. Normal pituitary function in this case (will be followed)

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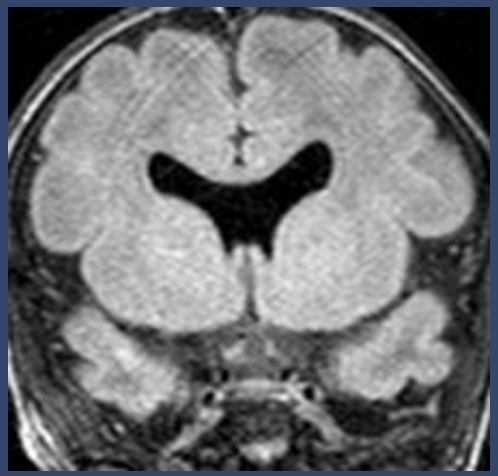
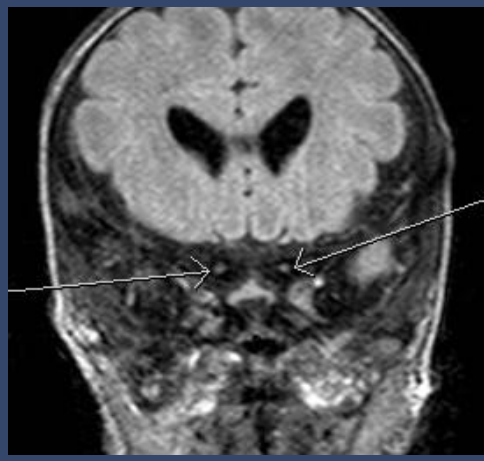
Septo-optic Dysplasia

- Findings (variable)
  - Hypoplastic Optic nerves/tracts
  - Absent CSP
  - Hypothalamic pituitary dysfunction
- Typical frontal horn morphology
  - Squared off horns in axial and coronal planes
  - Downward point to anterior horns of lateral ventricles on coronal plane
  - Fornices are **not fused**
    - **If fused, consider lobar holoprosencephaly**
- Resolution of MR and US not good enough to diagnose small optic nerves…YET?

Amirsys

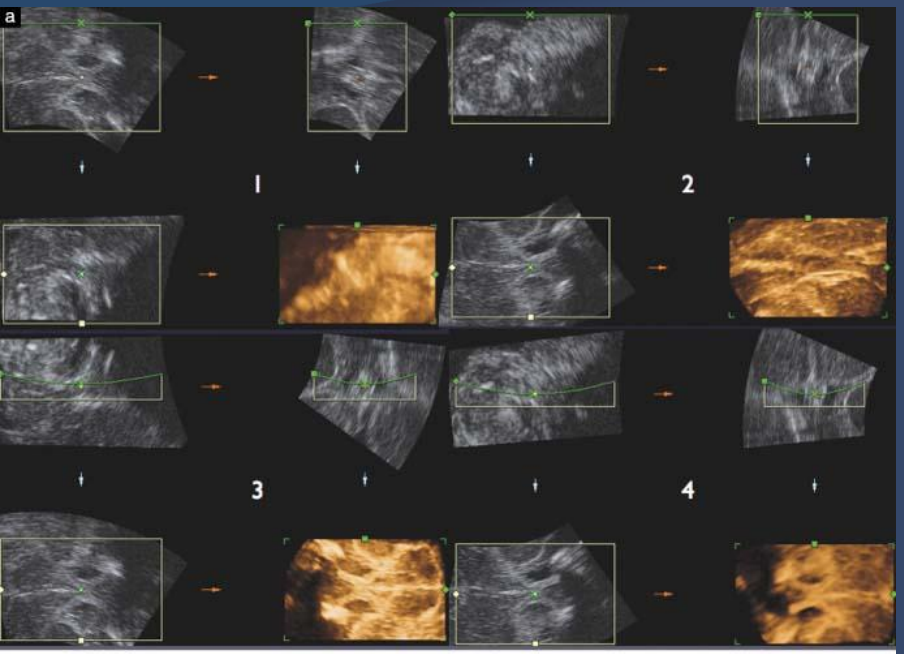


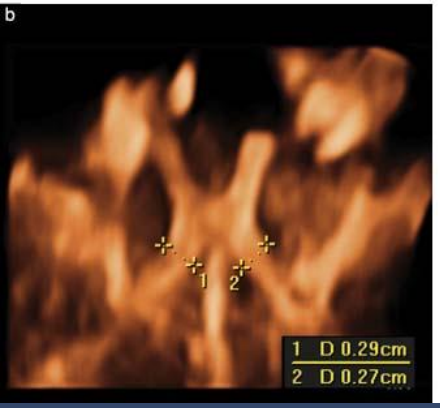


Role of three-dimensional ultrasound measurement of the optic tract in fetuses with agenesis of the septum pellucidum

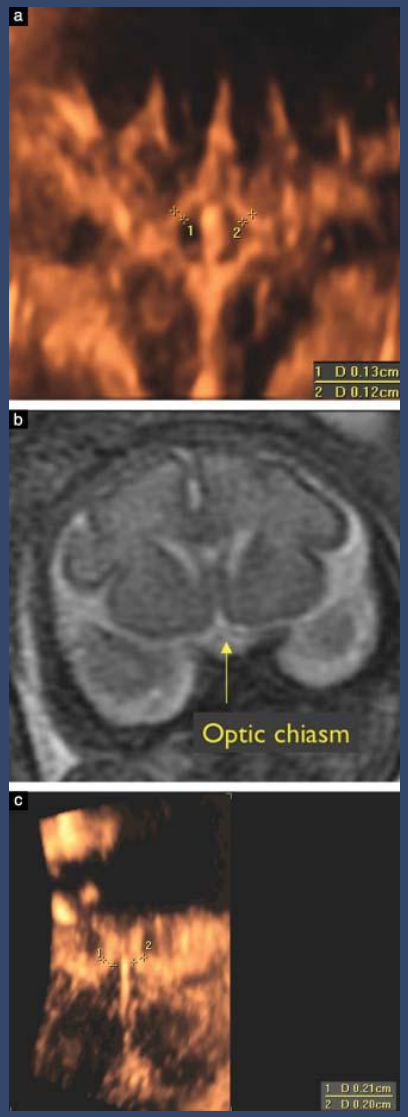
J. P. Bault[1,2,3,*], L. J. Salomon[2,3], L. Guibaud[4], R. Achiron

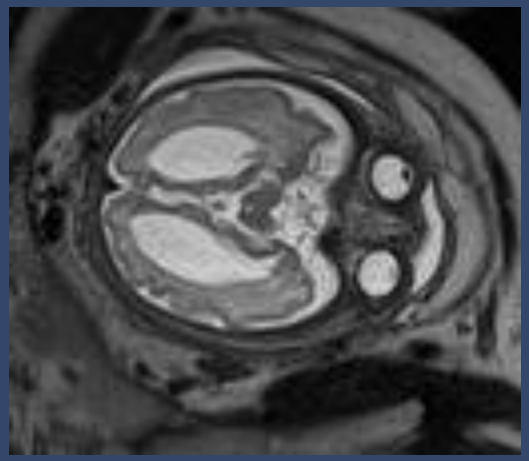
Ultrasound in Obstetrics & Gynecology, Volume 37, Issue 5, pages 570-575, 21 APR 2011 DOI: 10.1002/uog.8847





Normal Optic Chiasm





From Anne Kennedy
University of Utah

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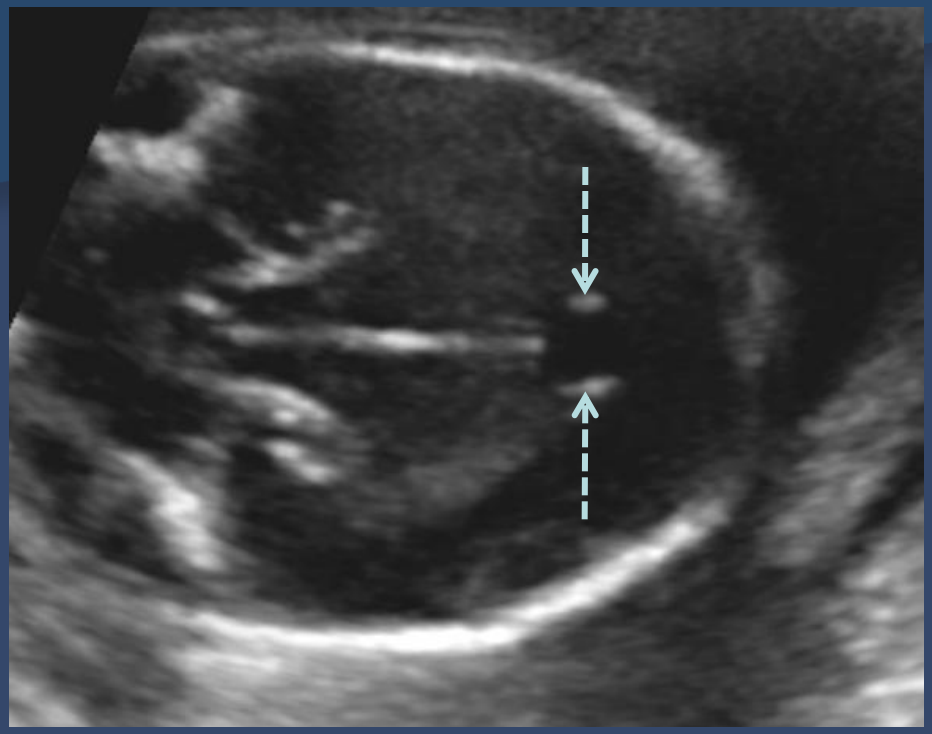
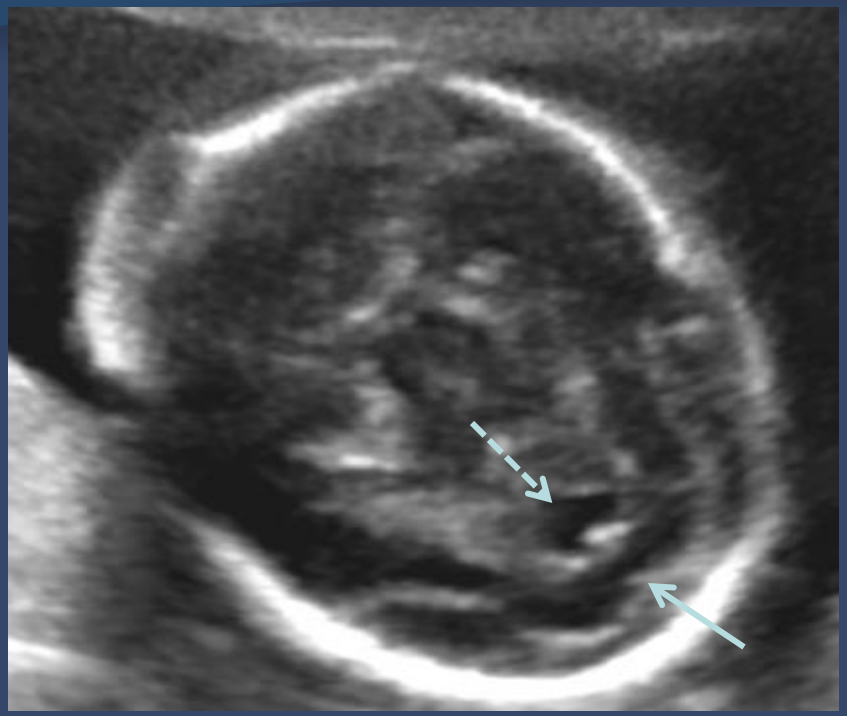
Case: Referral for absent CSP, 21 wk…
Is the anterior anatomy normal? Falx? Normal CSP?





- No Falx
- No CSP…CSP is a box flanked by frontal horns. In this case, no frontal horns! Box = Fused frontal horns (mimic CSP)
- Other findings:
  - Brain parenchyma crosses midline
  - Thalamus is not fused

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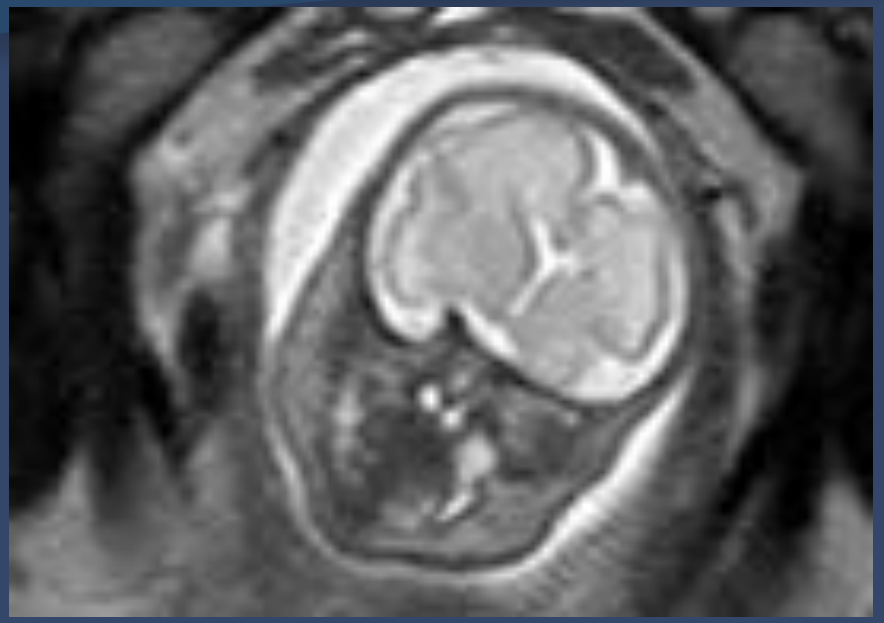
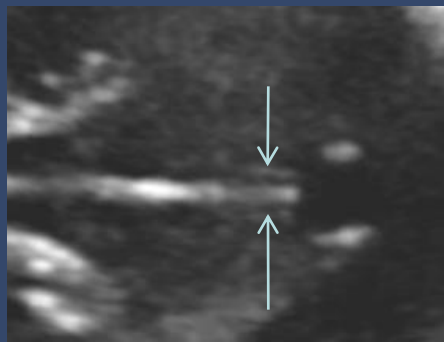
Case: Fetal MR



Lobar Holoprosencephaly

- Absent CC
- Cortex crosses midline anteriorly
- Frontal horns small and fused
- Fused fornices might help make dx
  - BUT not always fused



(OP14.04) Fused Fornices: CNS Findings and Outcome in 30 Fetal Cases

A. Kennedy MD[1], E. Deer MD[1], C. Nelson MD[2], K. Moore MD[2],

1:. Department of Radiology, University of Utah, Salt Lake City, UT, USA. 2: Primary Children's Medical Center, Salt Lake City, UT, USA

Introduction

Fused fornices has been proposed as a specific sign of lobar holoprosencephaly on prenatal imaging. We describe 30 cases of fused fornices identified on prenatal ultrasound which, on postnatal MRI, do not demonstrate cortical continuity across the midline. We propose caution in assigning an imaging diagnosis of lobar HPE when midline continuity of the cerebral cortex cannot be confirmed.

Methods

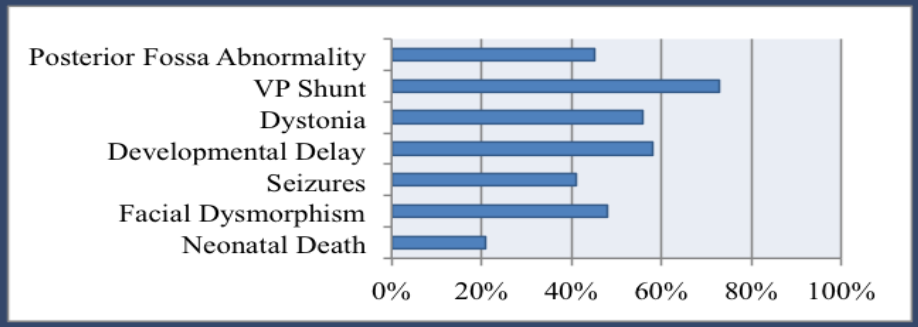
Prospectively we gathered 30 cases of fused fornices demonstrated on prenatal ultrasound. Postnatal MR was obtained in 24 patients. CNS and extra-CNS anomalies were tabulated with imaging and chart review.

Results

In addition to fused fornices, all patients exhibited the following features which can be associated with holoprosencephaly :

- Absence of the septum pellucidum
- Corpus callosum abnormalities
- Aqueductal stenosis with hydrocephalus

Figure 1. Fused fornices on prenatal (A), neonatal (B) ultrasound, and postnatal MR (C).



Conclusion: Forniceal fusion is not a specific sign of lobar holoprosencephaly. When found on fetal imaging, it should be considered a sign of a midline brain malformation with a high likelihood of concomitant CNS and extra-CNS anomalies and poor prognosis.

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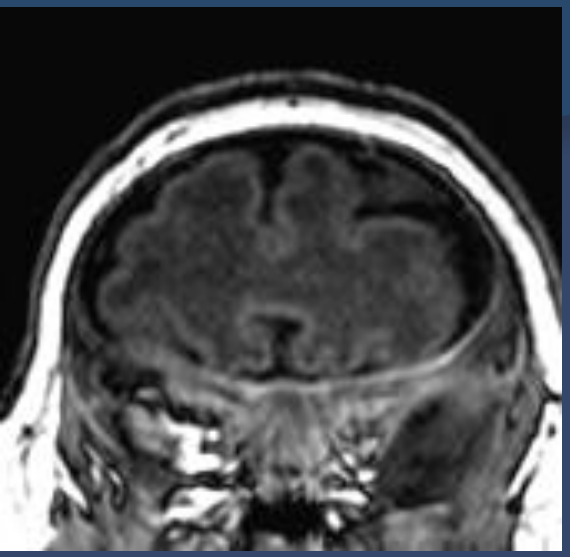
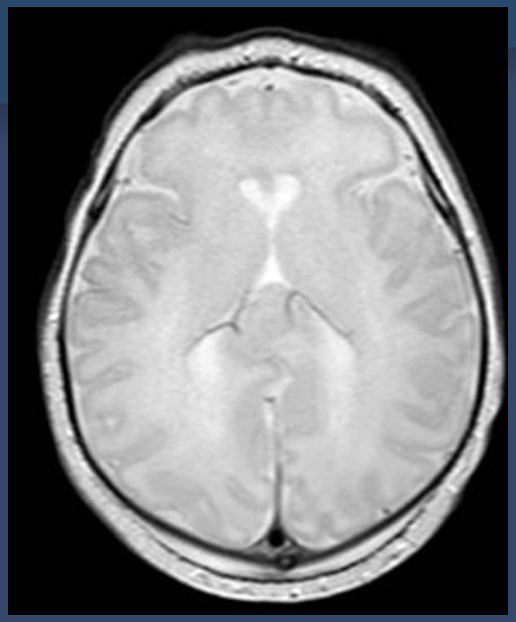
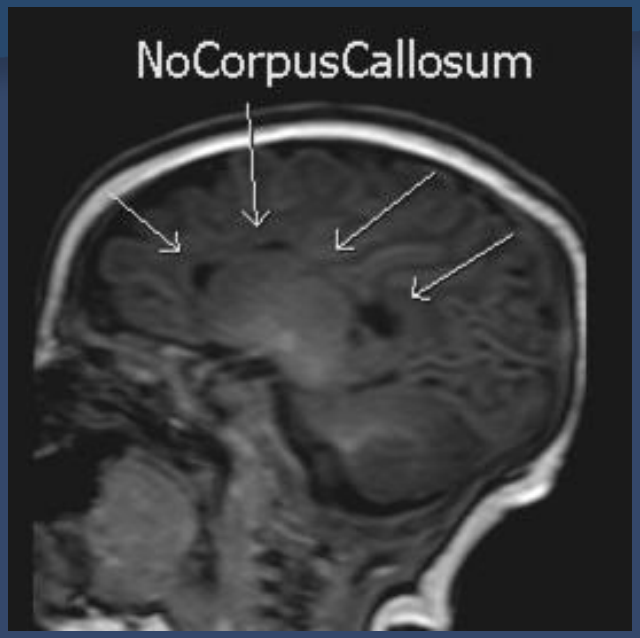
Baby MR: Lobar Holoprosencephaly







- Clinical findings (even if mild)
  - Microcephaly and facial anomalies
  - Severe cognition delay
  - Spastic quadriplegia
  - Visual disorders
- Associated with aneuploidy (T13)

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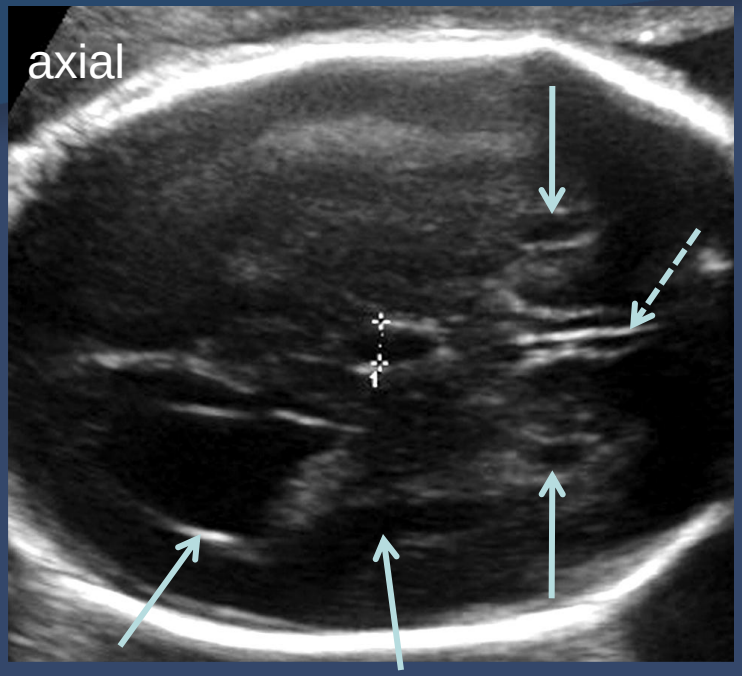
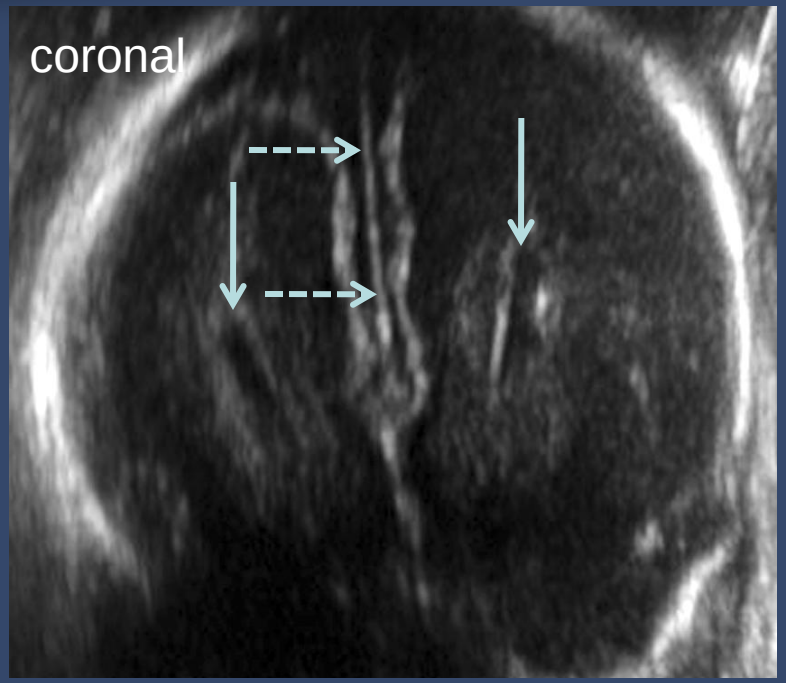
Case: Falx? CSP? Are the frontal horns normal?





- Falx? Abnormal…extend too far posterior/inferior
- CSP? No
- Vents: teardrop shaped, parallel frontal horns
- Diagnosis: Agenesis of Corpus Callosum

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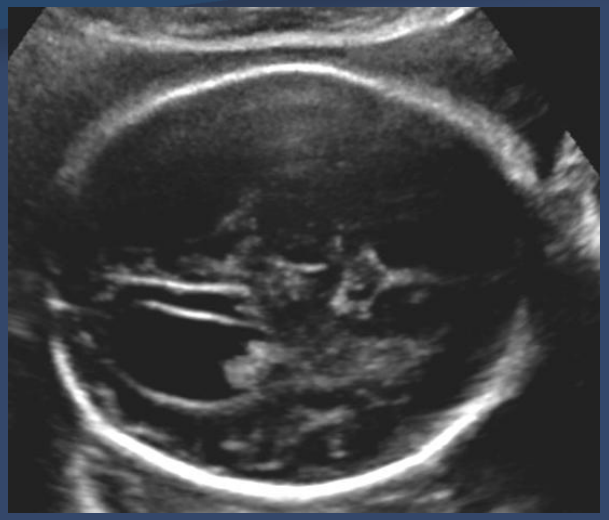
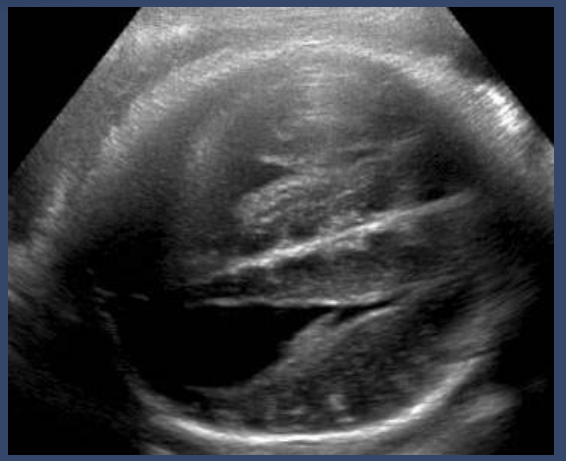
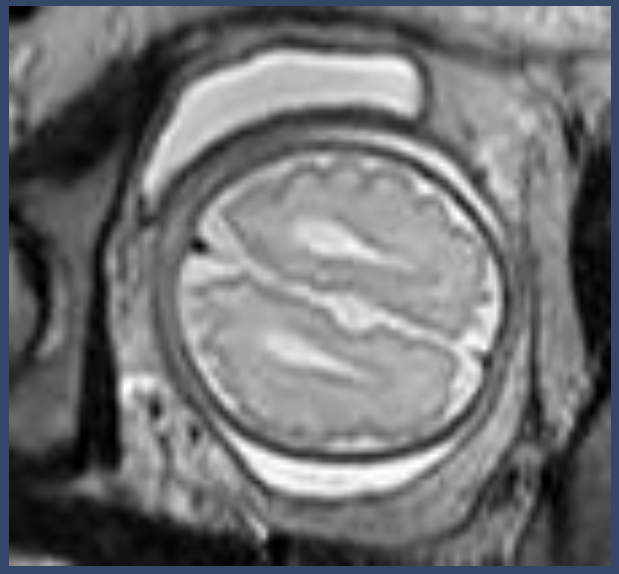
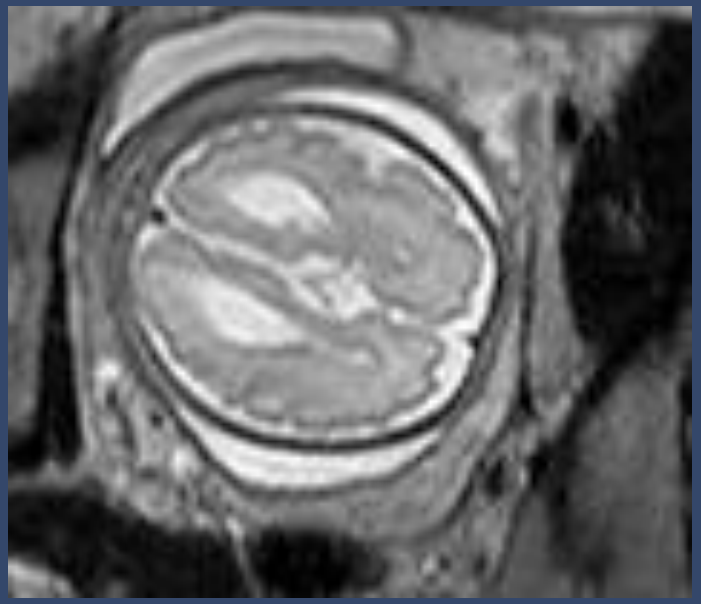
A key finding on axial view with agenesis of corpus callosum

Colpocephaly : lateral ventricle bulge in back, narrow in front









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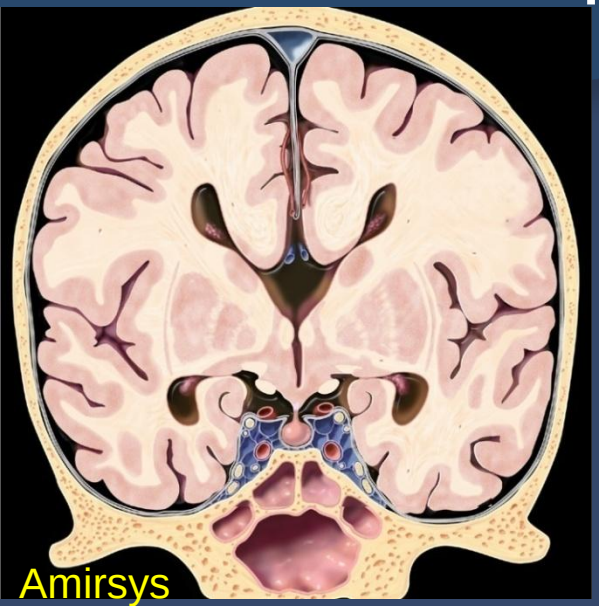
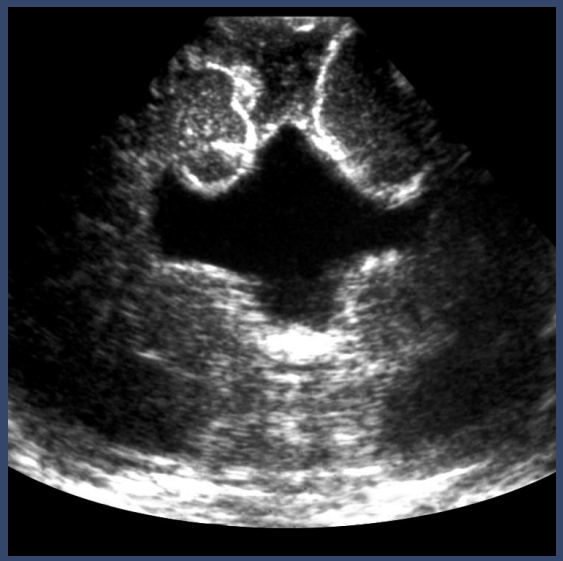
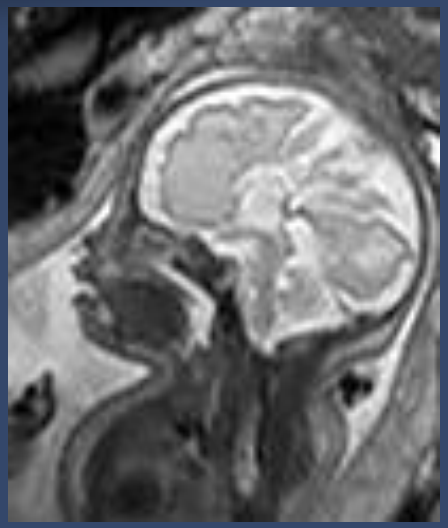
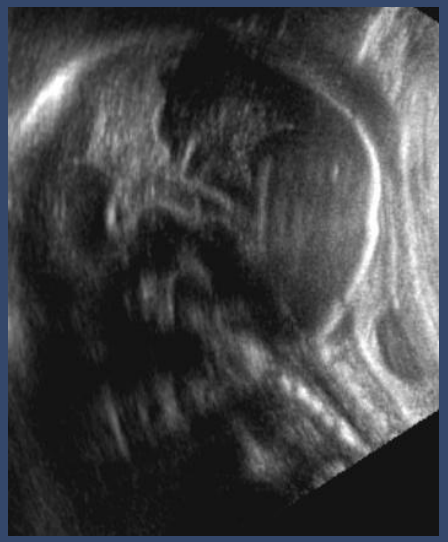
Agenesis of CCL: Unique coronal anterior appearance
Absent CSP and rise of 3rd ventricle
"Texas longhorn" "Trident" shape or "Trident"





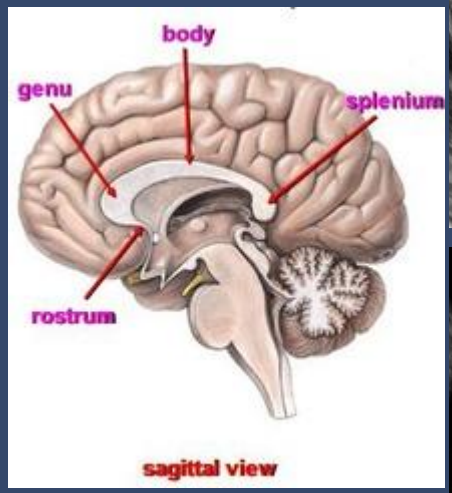
Sagittal plane shows absence of CC and radial orientation of medial gyri

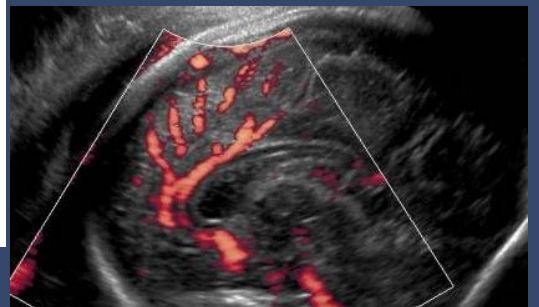




Classic teaching: “CC forms from Front to Back”

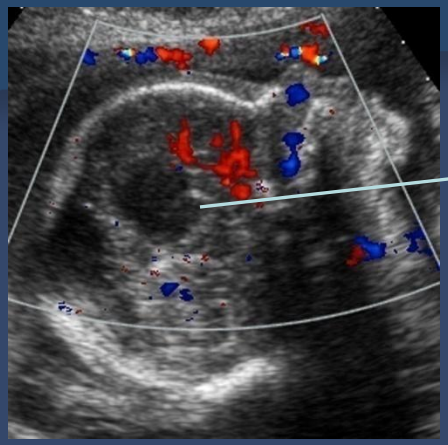
In reality might have bits and pieces:
Posterior genu/anterior body THEN anterior genu/posterior body followed by splenium and rostrum
Pericallosal artery follows the CC

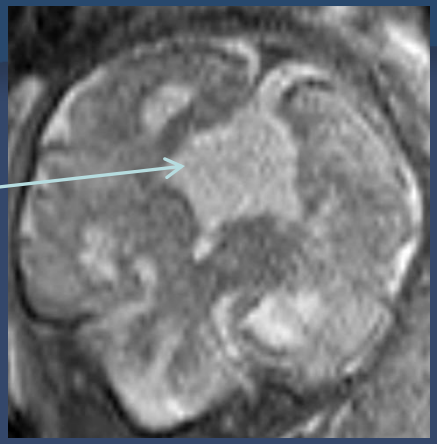


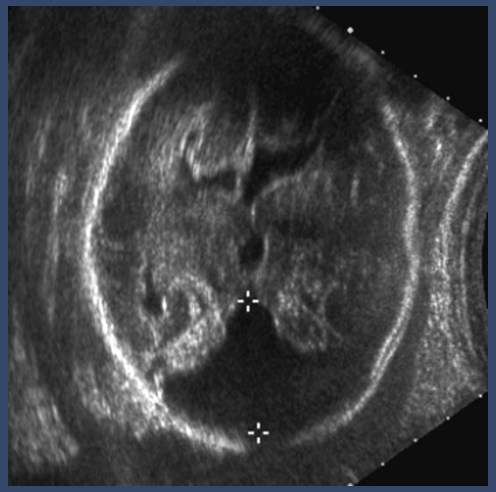


Agenesis of the Corpus Callosum

- Diagnosis by Pattern Recognition
  - Absent CSP
  - Parallel frontal horns
  - Elevated 3rd ventricle
  - Colpocephaly of LV
- Other CNS anomalies (not subtle)
  - Central cysts
  - Lipoma
  - Cerbellar vermian defects
- Non CNS anomalies in 60%
- Aneuploidy in 10-20%
- Syndromes in 50-80%
  - Autosomal dominant, recessive and X-linked described







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Suggested reading

The Cavum Septi Pellucidi

Why Is It Important?

Thomas C. Winter, MD, Anne M. Kennedy, MBBCh, Jan Byrne, MD, Paula J. Woodward, MD

J Ultrasound Med 2010; 29:427–444

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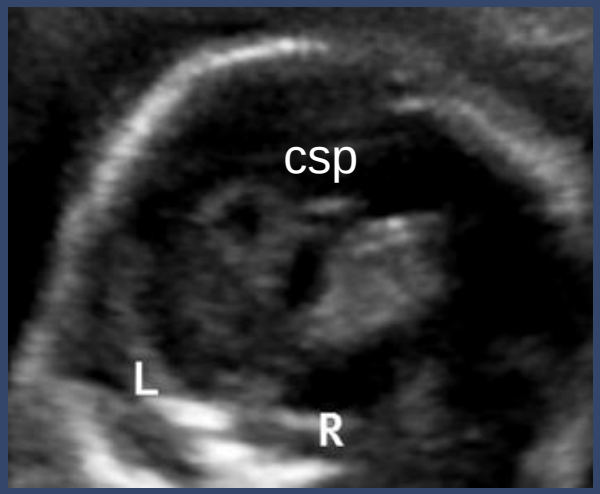
Ventricles
Case: 23 wk 1 wk s/p laser for TTT
Are the ventricles normal? Is CP normal?



- Vents are not dilated.
- Echogenic material in anterior right lateral ventricle with inreased wall echogenicity
- Remember no choroid in LV “horns”:
  - frontal, temporal, occipital horns
- Diagnosis: Intraventricular hemorrhage, Grade 2

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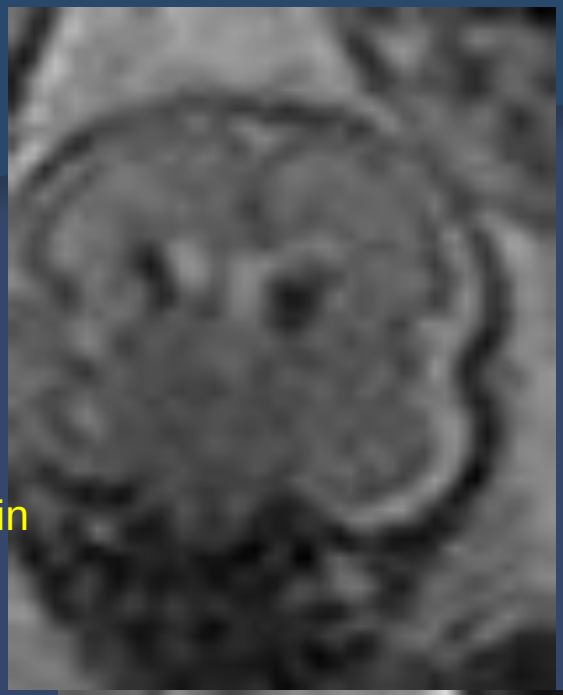
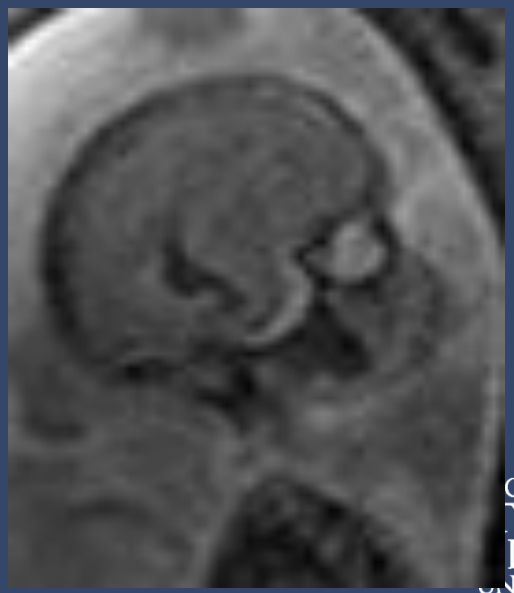
Fetal MR in same case : Bilateral hemorrhage

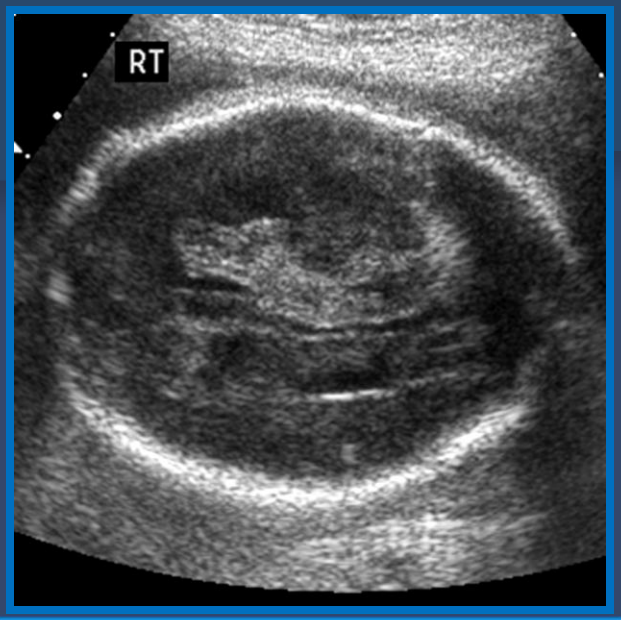
T2 weighted MR
Low signal = hemosiderin

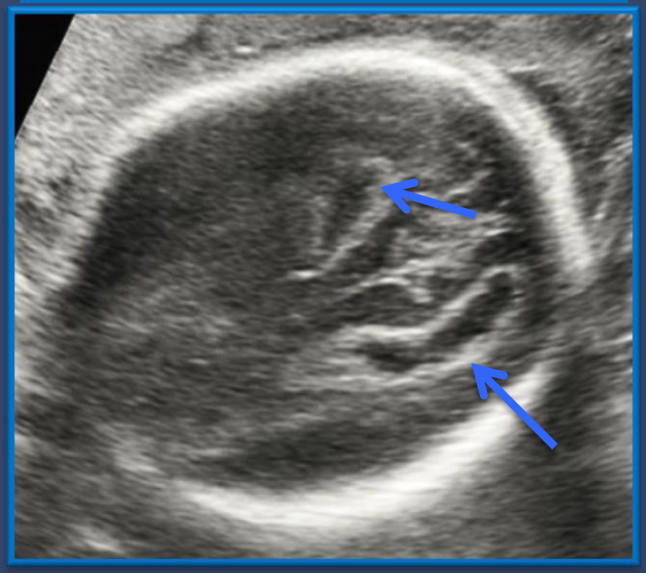


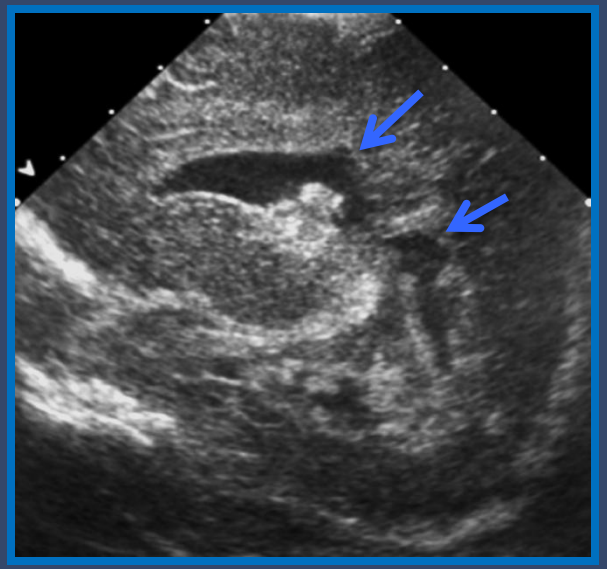


Hemorrhage: Subtle Findings

- Loss of normal cerebral landmarks
- Hyperechoic, avascular intracranial mass
- Hyperechoic acute clot adherent to “bulky/nodular” choroid plexus
- Hyperechoic clot outlining cerebral cortex
- Hyperechoic nodular ependyma
- Increased periventricular white matter echogenicity
- Ventriculomegaly
- Porencephaly







McKlean, Kennedy,Woodward
Reference, RSNA 2011

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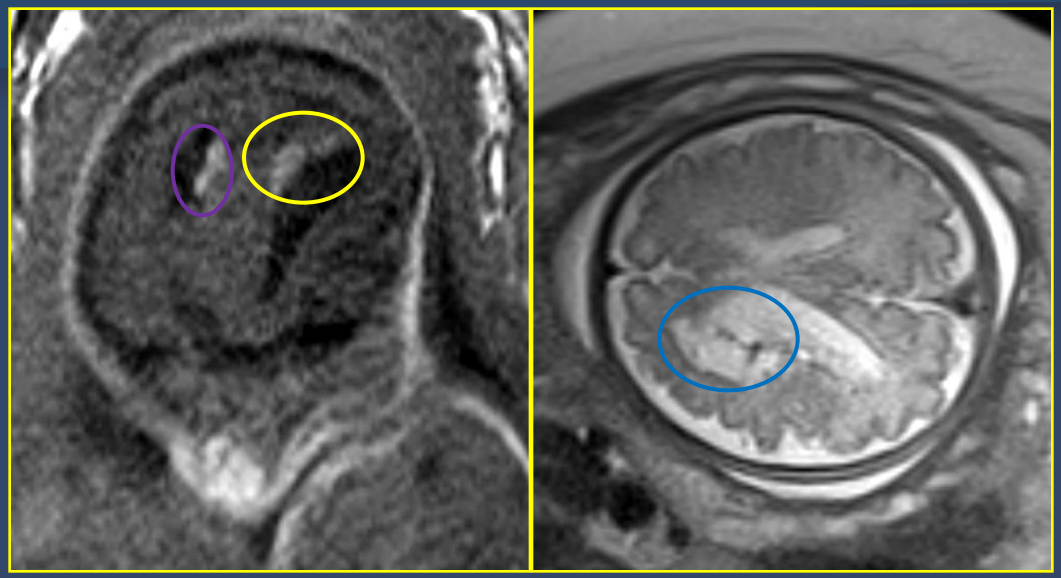
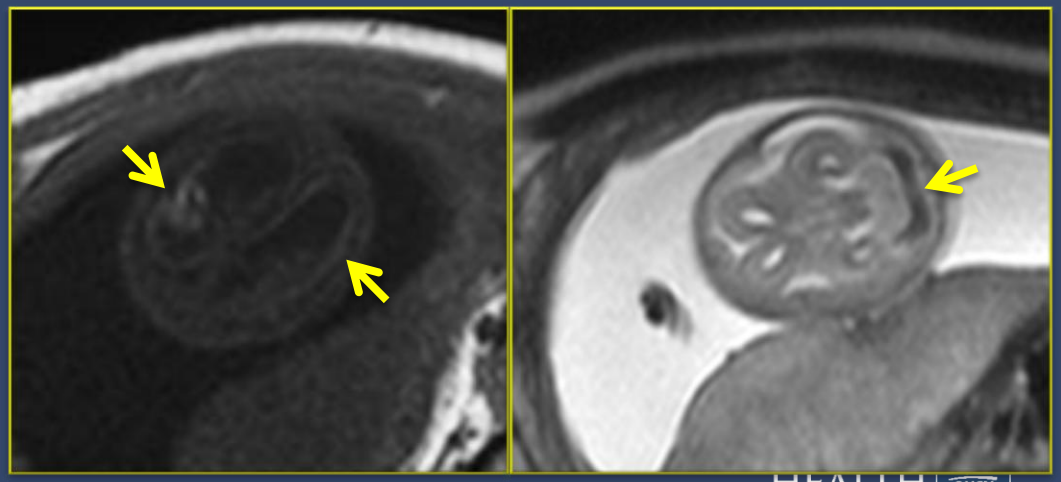
MRI findings

- T1 WI: High Signal
  - High signal clot
  - High signal parenchymal foci suggest vasculitis or petechial hemorrhage
- T2WI: Low signal
  - Low signal clot
  - Fluid/fluid level
  - Porencephalic cyst (high signal)
  - Abnormal cortical signal





McKlean, Kennedy,Woodward Reference, RSNA 2011

Fetal Hemorrhage: Grading and Outcome

Neonatal Grading

- Grade 1
  - Germinal matrix only
- Grade 2
  - GMH + Ventricular blood
- Grade 3
  - Grade 2 + Ventriculomegaly
- Grade 4
  - Parenchymal hemorrhage
- Prognosis:
  - Excellent for 1and 2
  - Variable for 3
  - Poor for 4

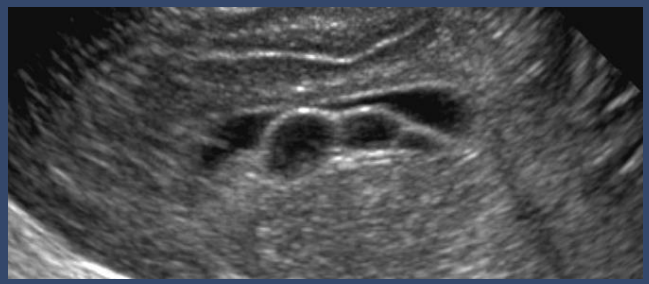
Fetal Dx outcome

- IVH grade 3-4 with poor outcome
- Grade 1 with good outcome
- Grade 2 is variable
- All hemorrhages: 47% normal outcome at 12 months

References:
Elchalal U, Ultrasound Obstet Gynecol 2005 sep 26(3)
Ghi T, et al; Ultrasound Obstet Gynecol 2003 aug; 22 (2)

Other subtle lateral ventricle anomalies: Cysts

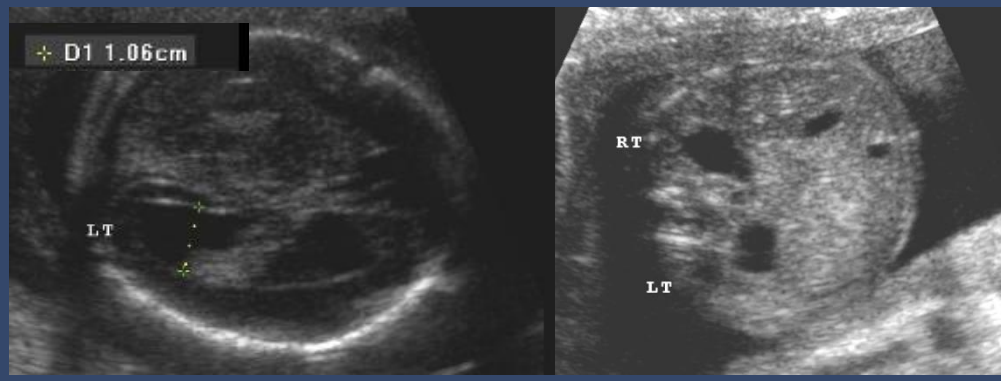
- Connatal cysts
  - Frontal horn entrapment “cysts.
  - DDX: old hemorrhage
- Ependymal cysts
  - Occur anywhere there is ependymal cells
  - Can cause obstruction



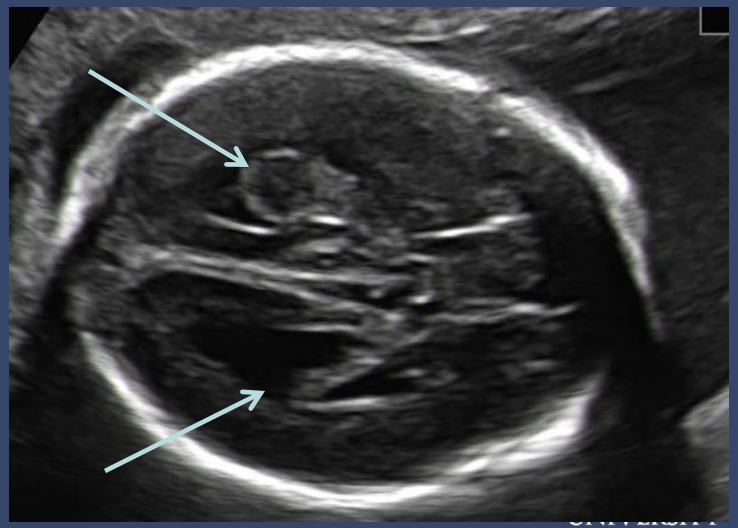
FINALLY, Isolated Mild Lateral Ventricle Dilatation

- When isolated it's a marker
  - Not an anomaly
- DDX:
  - Normal (80%)
  - Aneuploidy
  - Beginning of something worse
    - Hydrocephalus
  - As part of another anomaly
    - Consider MR
- Definition:
  - 10-12 mm (others use 10-15)
  - Measure it right!

MILVD + pelviectsis: outcome T21



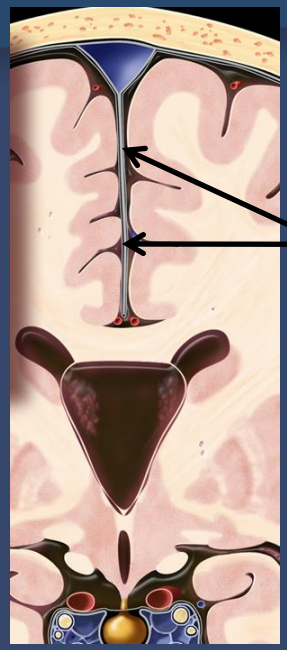
MILVD + CPC , outcome NORMAL



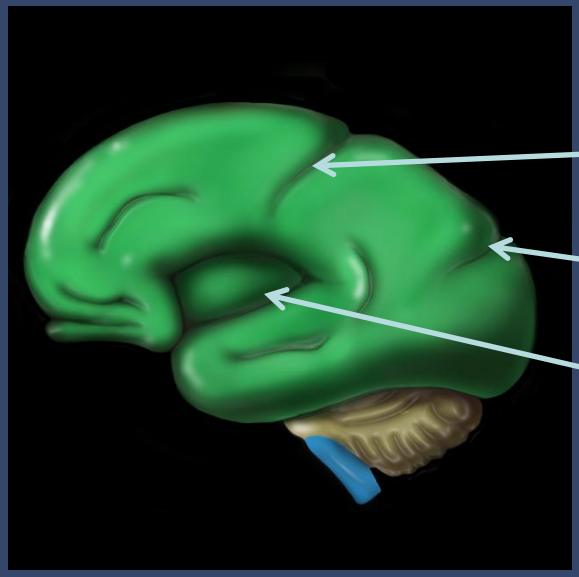
Websites with good information on ventriculomegaly:
http://www.childrensmemorial.org/depts/fetalhealth/ventriculomegaly.aspx
http://fetus.ucsfmedicalcenter.org/ventriculomegaly/
http://prenatalpediatrics.org/vent.html

Shifting Gears: Parenchymal anomalies

- Notice fissures and Sulci
- Cortical anomalies
  - Lissencephaly
  - Schizencephaly
  - Gray matter heterotopia
  - Pachygyria/polygyria



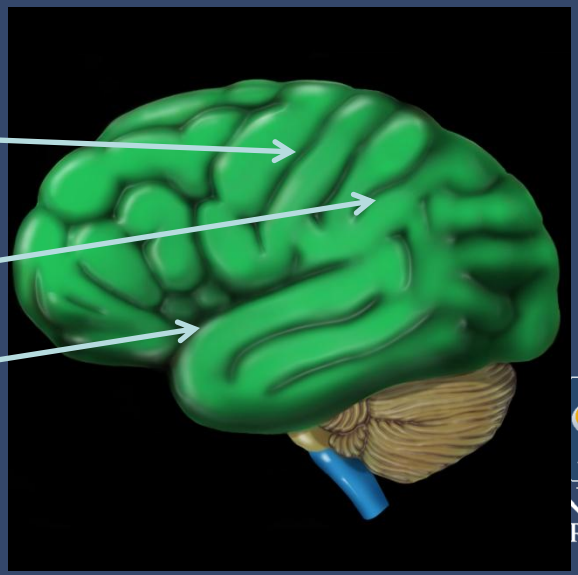
Amirsys



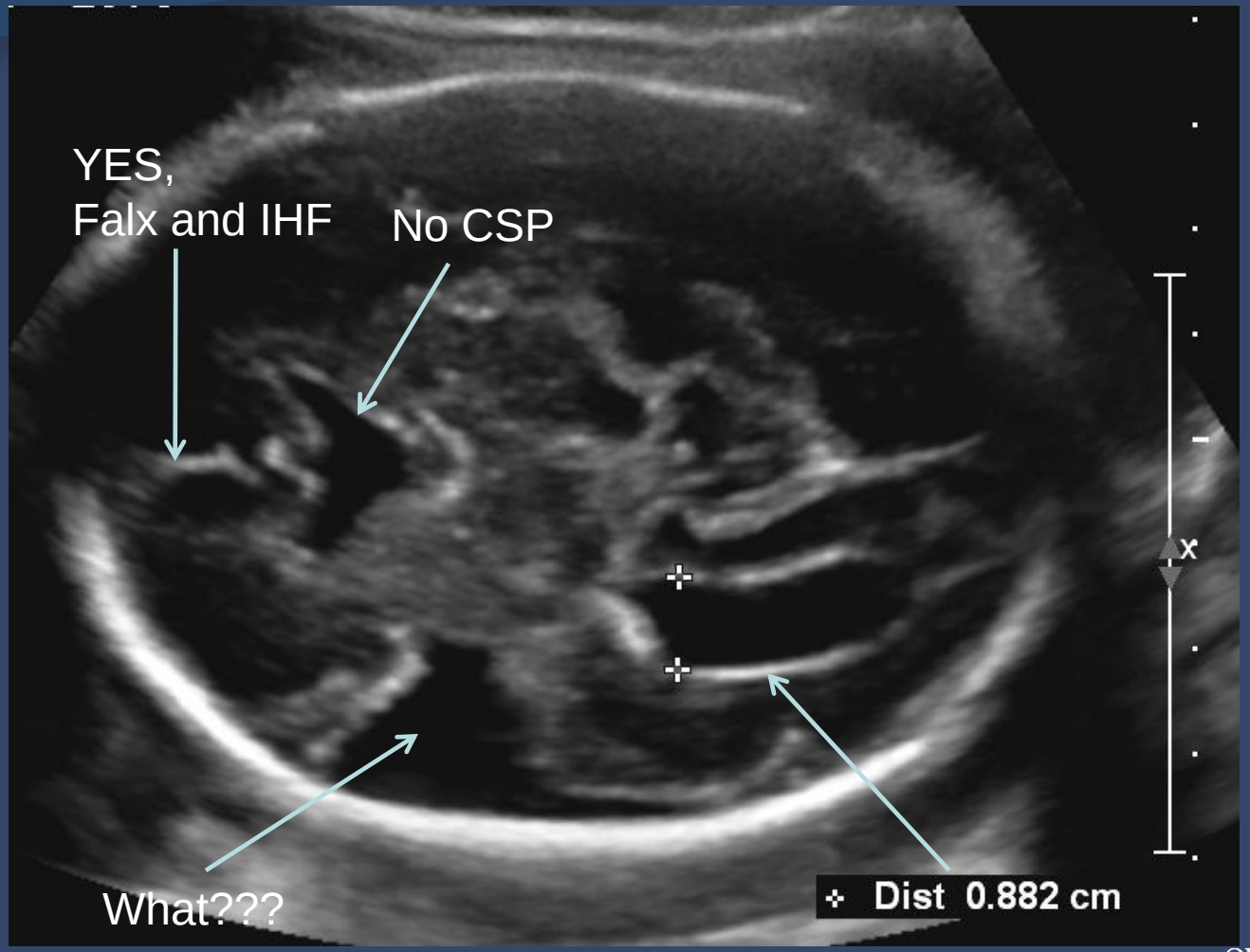
Central sulcus

Parieto-occipital sulcus

Sylvian fissure



Case: referral for absent CSP
Is there a falx? CSP? Vents? Sulci and fissures?



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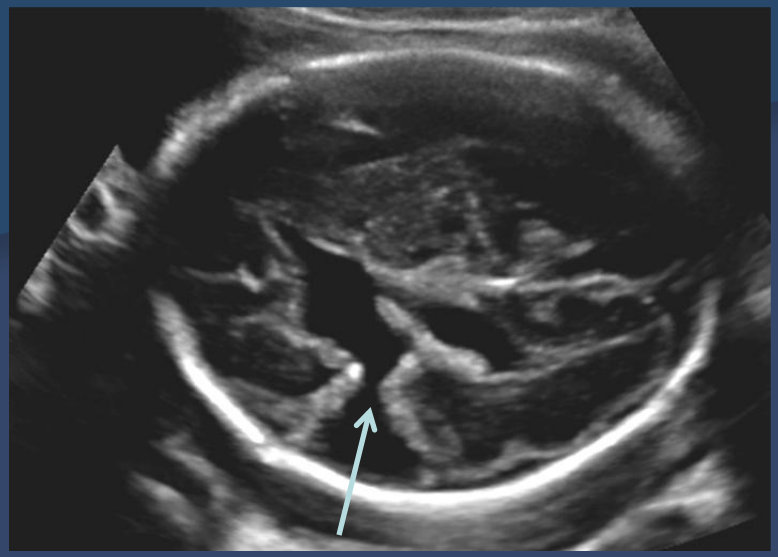
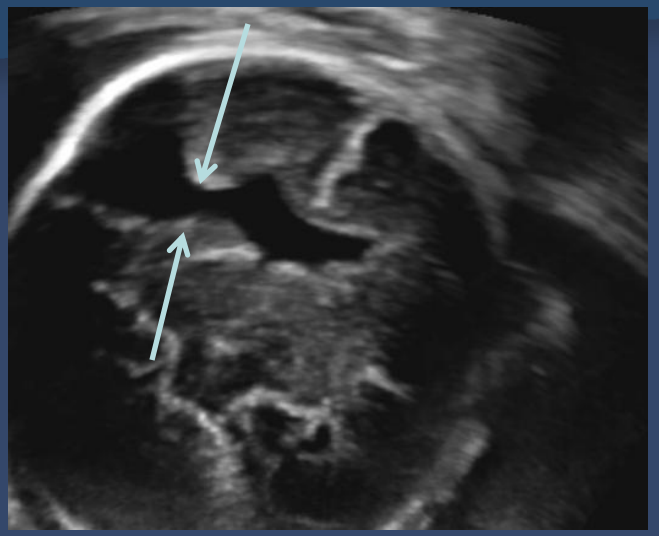
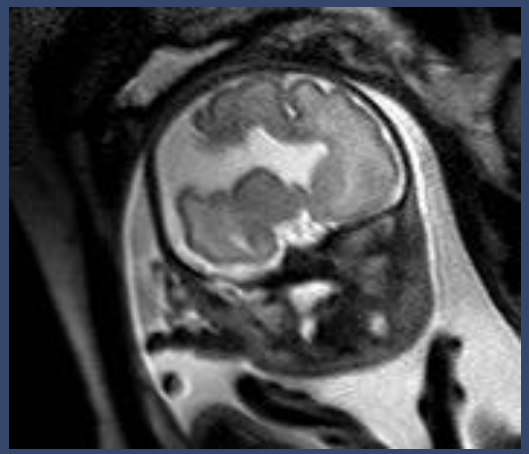
Additional non-standard views and MR







Diagnosis: Schizencephaly

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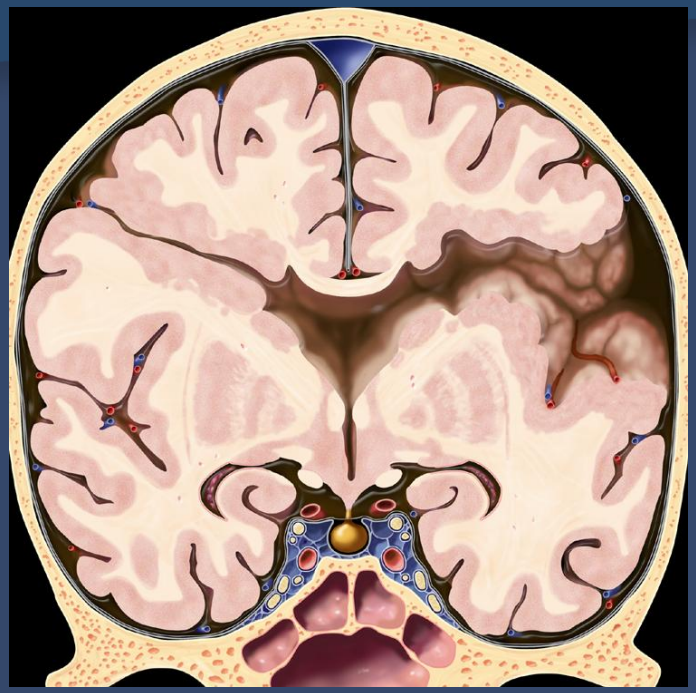
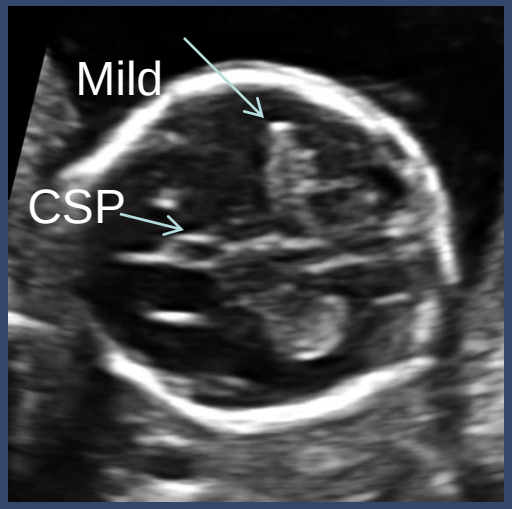
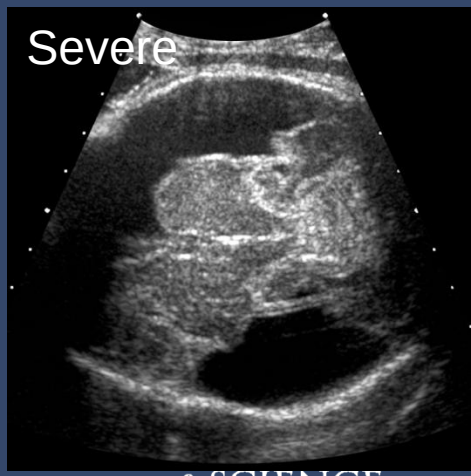
Schizencephaly

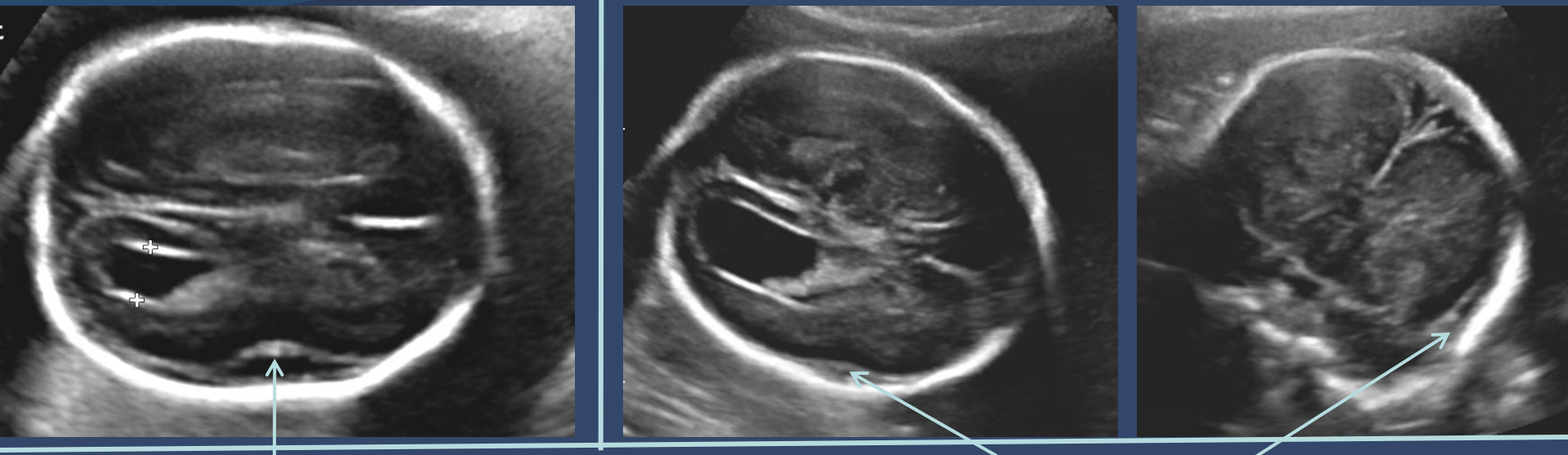
- Brain parenchyma cleft lined by gray matter extending from skull to ventricle
  - Closed-lip
  - Open-lip
- Neuronal migrational anomaly vs early prenatal injury
  - Unilateral in 60%
  - bilateral in 40%
- Imaging findings
  - Variable
    - Mild: Tenting of vent wall
    - Severe: Large open defects
  - CSP absent in 70%
  - MR detects other subtle cortical anomalies
    - Mirror image distribution







Case: 23 wk mild ventriculomegaly progressed at 28 wks and sent for referral



23 wk, sylvian fissure

28 wk

- Progressive ventriculomegaly
- Smooth brain
- Suspicion: Lissencephaly

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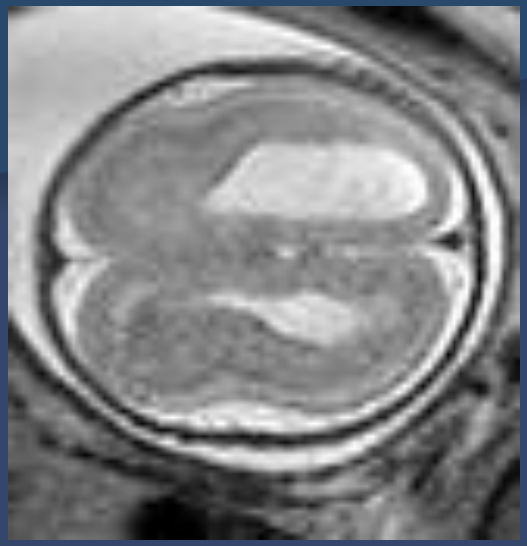
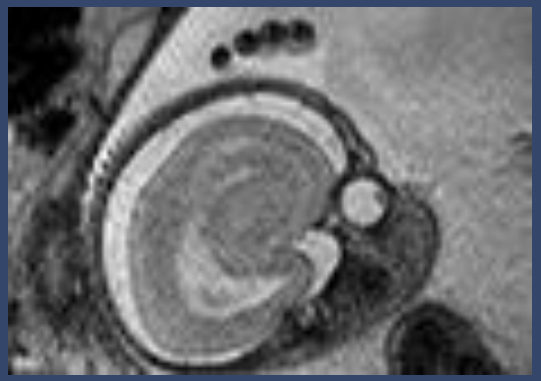
MR: 28 wk





- Brain sulcation pattern of 20 wk fetus: shallow sylvian fissures, no definition of parieto-occipital sulcus. disorganized cortical gray matter.
- DX: Lissencephaly and heterotopia
- Baby was Dxd with Miller Dieker syndrome

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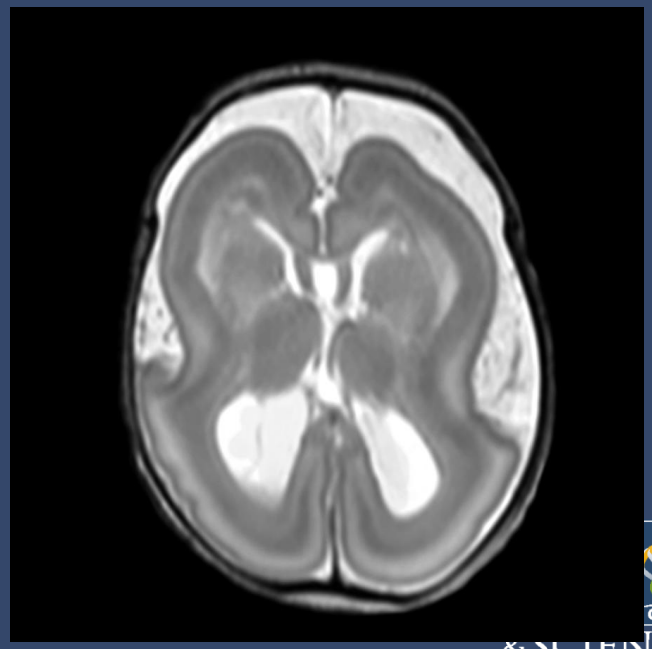
Lissencephaly

- Abnormal smooth brain surface
  - Isolated or part of syndrome
  - Miller Dieker syndrome
  - Walker Warburg
- Associated with other cortical defects
  - Polymicrogyria: many small gyria
  - Pachygyria: broad flat gyria
- Band heterotopia common:
  - Thick band like gray matter

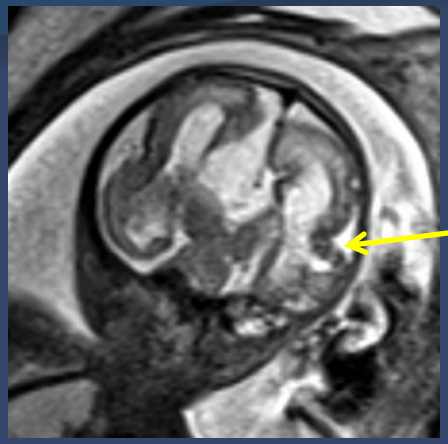


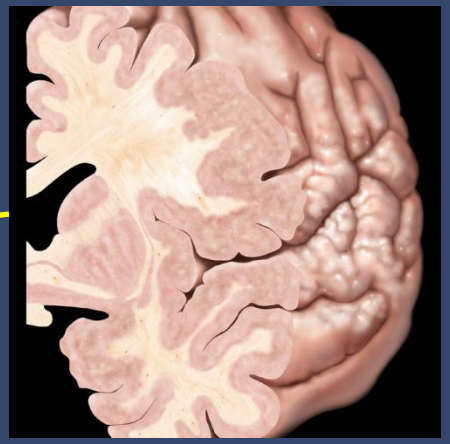
Structure	US	MR
Callosal sulcus	14	22
Sylvian fissure	18	24
Parieto-occipital fissure	18	22-23
Calcarian fissure	18	22-23
Cingulate sulcus	24-26	28-29
Central sulcus	26	26-27
Convexity sulci	28	28-29

AMIRSYS cases, table and diagram

Pachygyria and Polymicrogyria diagnosis

Typical sequence: US suggests lissencephaly and MR shows more





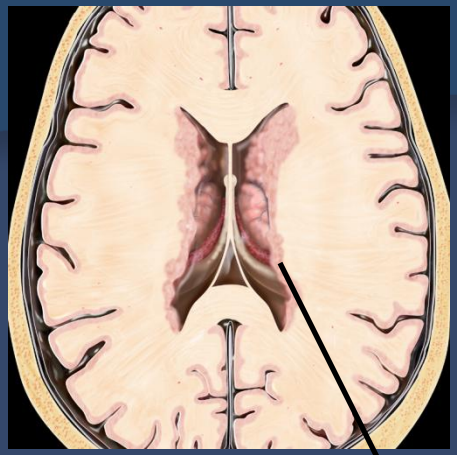
Polymicrogyria

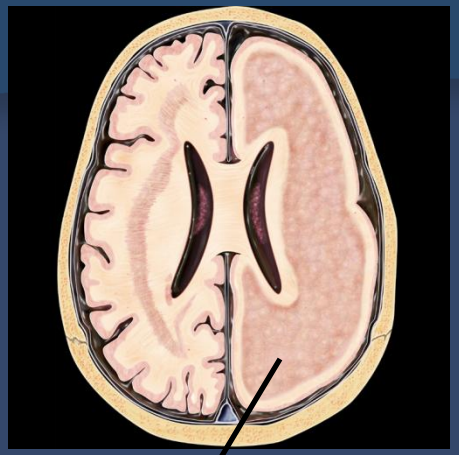
Pachygyria: Large broad based, flattened gyria

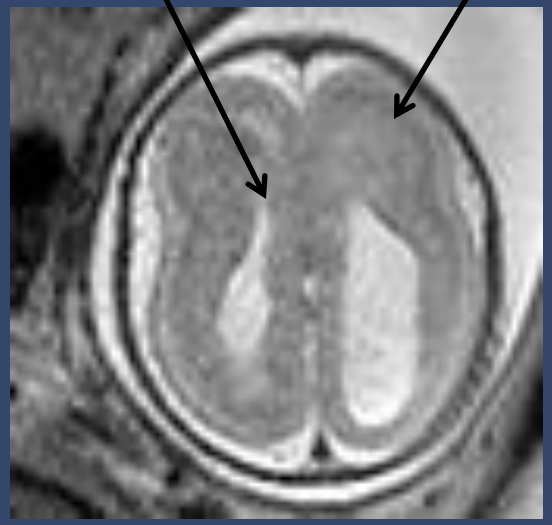
AMIRSYS cases and diagram

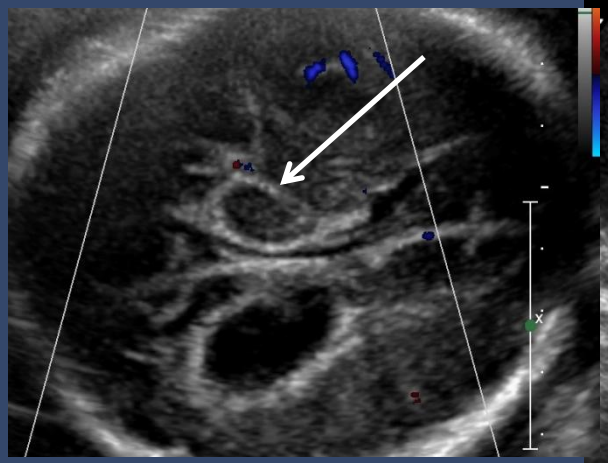
Gray matter Heterotopia

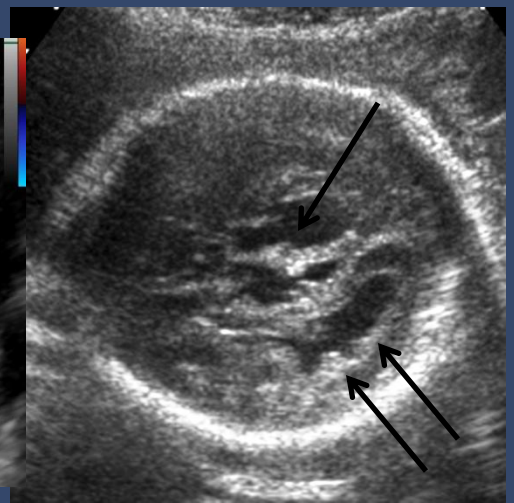
- Abnormal nerve cells in nodular or laminar pattern in locations other than cortex
- Imaging features:
  - Mild with nodular ventricular wall
  - In association with other cortical defects
    - Band heterotopia and lissencephaly











AMIRSYS diagrams

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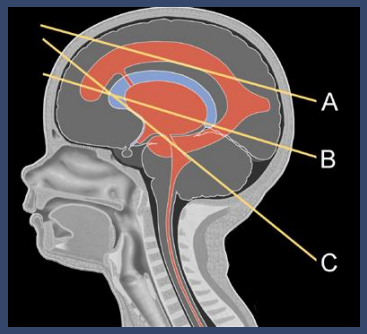
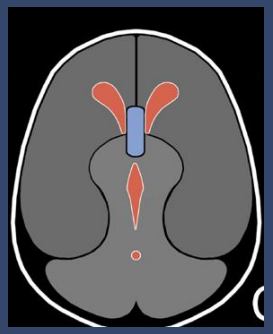
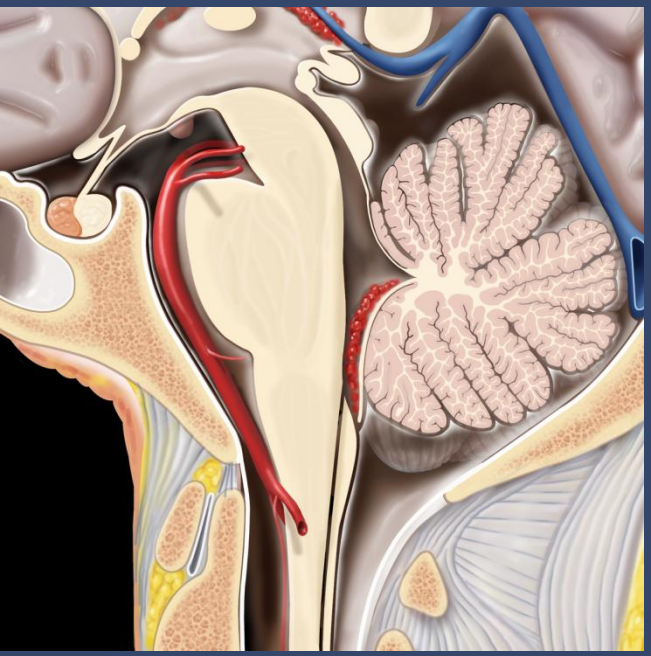
Shifting gears: Posterior Fossa

- One routine view: Axial angled through cerebellum
  - Bi-lobed cerebellum, intact vermis, +/- dural folds, nuchal skin, intact calvarium
- 3D, Coronal and Sagittal if axial is abnormal
- TVUS if cephalic
- MR

Standard View







Amirsys drawings

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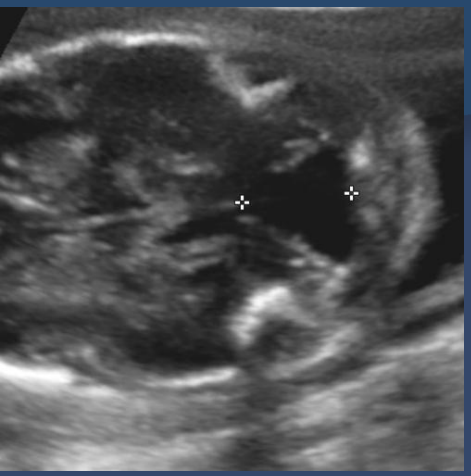
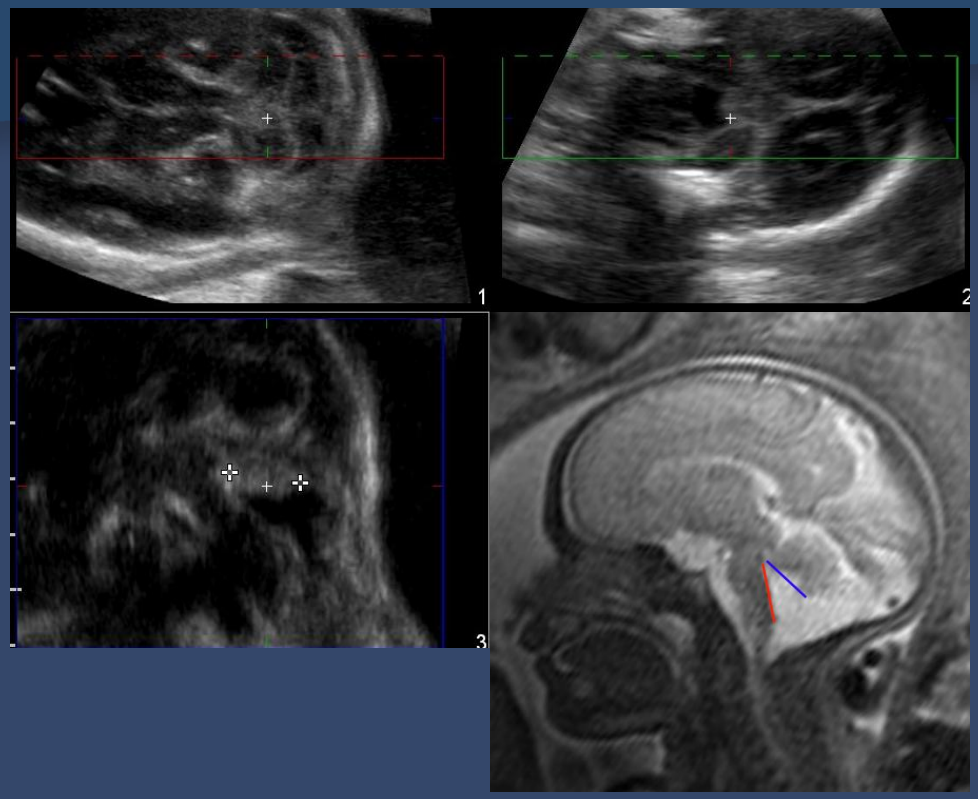
Case: Posterior fossa cyst: Is the vermis normal





Blake pouch cyst

- Posterior ballooning of superior medullary velum into cisterna magna (lifts the fastigial point of fourth ventricle)
- On axial imaging looks like cyst communicates with 4th
- Sagittal shows cyst is inferior to fourth and vermis is normal
  - Abnormal tegmentovermian angle
- Associated with ventriculomegaly
- Excellent outcome if isolated

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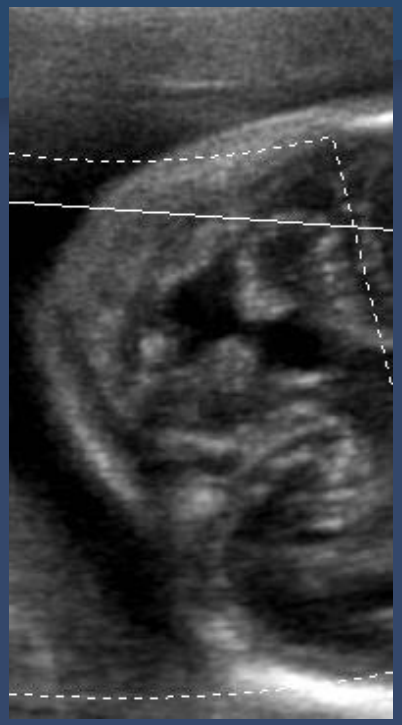
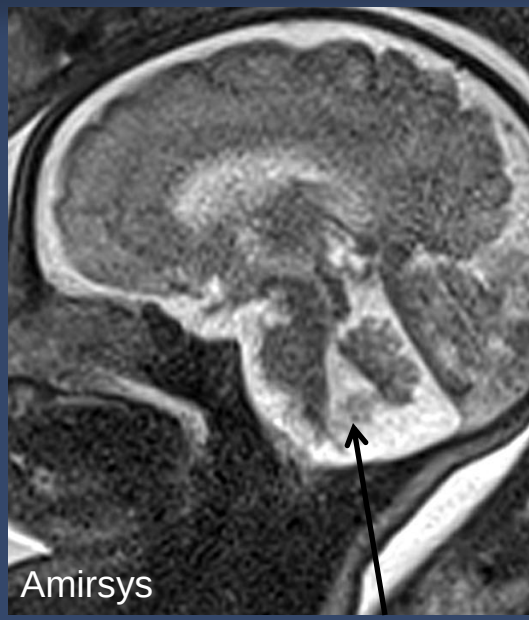
Case: Posterior fossa cyst. Is vermis normal?



Amirsys



Beware medial rotation of cerebellur

Inferior Vermian agenesis: partial or complete

- "Dandy-Walker variant" part of "Dandy-Walker Continuum"
- Sagittal view is key
  - Vermis should cover 4th ventricle OW there is communication between CM and 4th
- Axial: Key-hole appearance: dilated 4th.
- Beware cerebellar rotation: may mimic inferior vermis defect
- Beware early diagnosis: vermis forms until at least 18 wks!!!!

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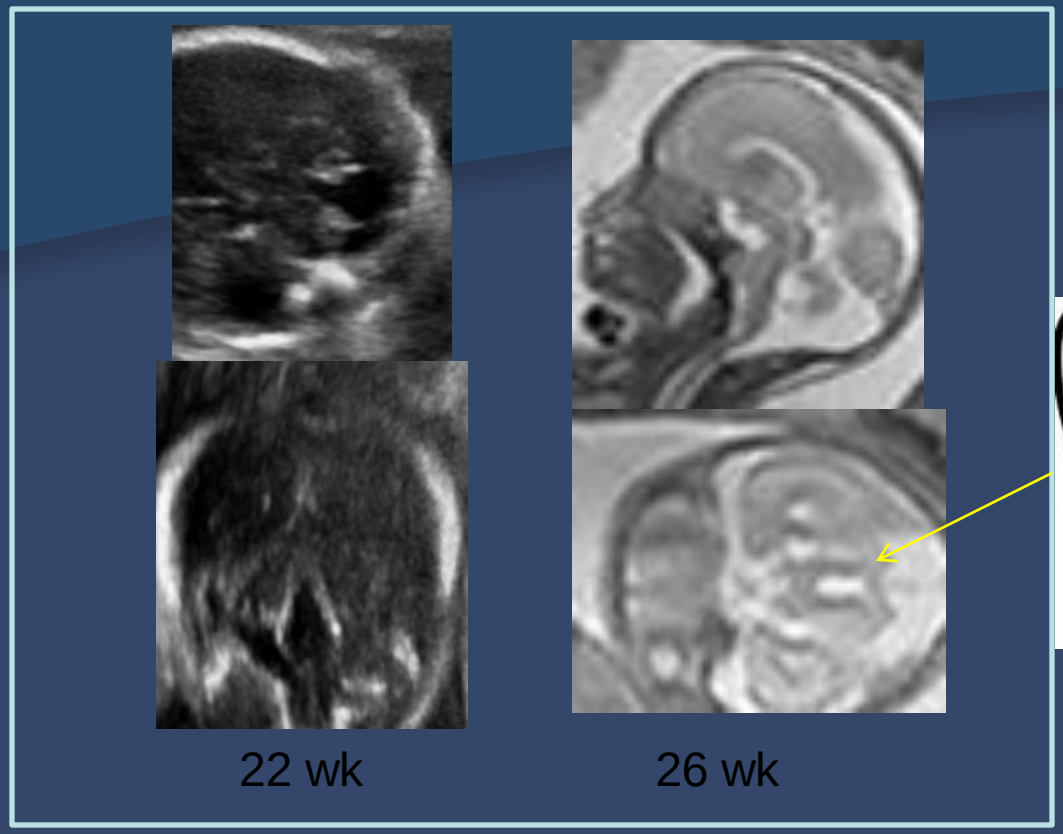
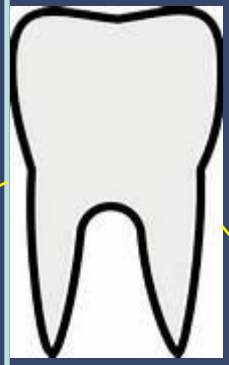
Cerebellar cyst : Joubert syndrome





- Vermis: small or absent
- 4th ventricle: large
- Key feature is “Molar Tooth Sign”: Elongated superior cerebellar peduncles

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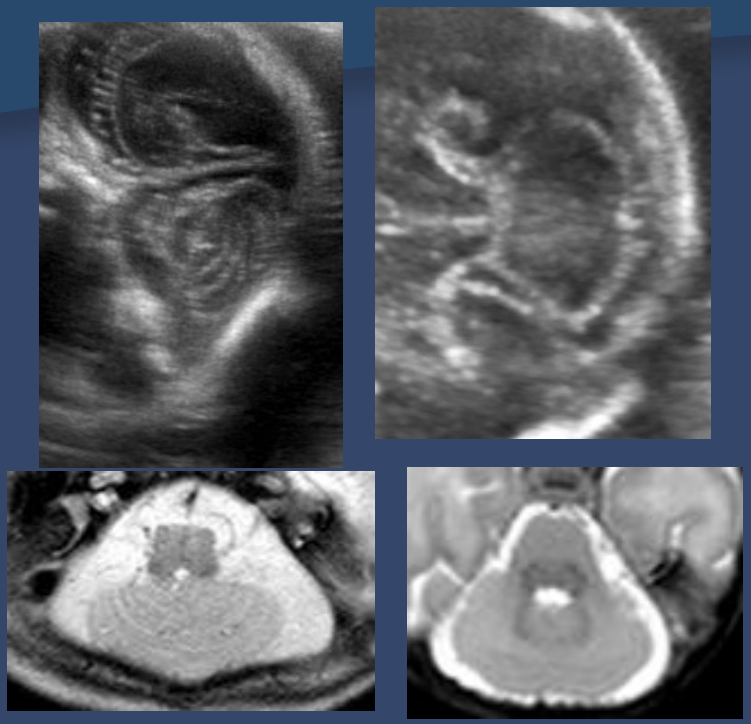
Cerebellar Morphology

Cerebellar hypoplasia



Rhombencephalosynapsis

Chiari 2

- Cerebellar hypoplasia
  - Measure cerebellar diameter (it's not always mega cisterna magna)
- Rhombencephalosynapsis
  - Fused continuous horizontal fovia
- Chiari 2
  - Cerebellar compression (not always banana sign)

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Summary

- Routine brain views are excellent for screening
- Absent cavum septi pellucidi is rarely an isolated finding, take it very seriously.
- Presence of the CSP is reassuring
  - "virtually ensures normal development of the central forebrain"

    -Dr. Roy Filly
- Isolated mild ventriculomegaly is common and often a benign finding (rule out everything else)
- Abnormal ventricular morphology suggests abnormal parenchymal morphology
- Approach to posterior fossa is strongly guided by morphology of vermis.
- When in doubt, get an MR. Even early MR is helpful