AMSER Case of the Month
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1 day old infant born at 34w3d with h/o hydrocephalus on prenatal US & pregnancy complicated by Substance Use Disorder (SUD)

Benjamin Soares, MS4

Bindu N Setty, M.D.

Boston University Chobanian & Avedisian School of Medicine

BU Chobanian & Avedisian School of Medicine

AMSER

Patient Presentation

HPI: Female neonate born via normal spontaneous vaginal delivery at 34w3d to a 19 y/o G3P0111 mother with recent hx of SUD (cocaine/fentanyl), gonorrhea/chlamydia (adequately treated), with US and MRI confirming the findings of severe dilation of lateral ventricles.

PE: Pre-term baby, moderately coarse breath sounds bilaterally, sacral dimple with no hair tuft/midline defects, decreased tone in upper and lower extremities, decreased Moro reflex, grasp reflex present, large head size with anterior fontanelle slightly depressed and a full posterior fontanelle.

Additional findings: Spontaneous motions of bilateral upper extremities with concern for seizures vs. withdrawal.

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

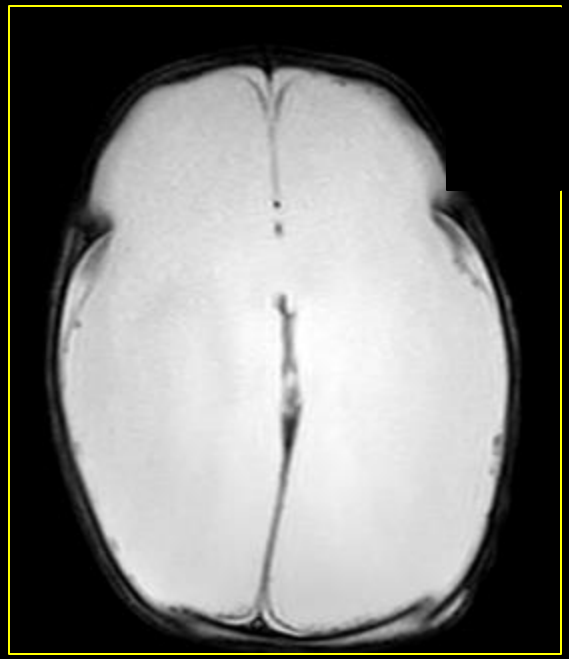
American College of Radiology
ACR Appropriateness Criteria®
Seizures-Child

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	☢☢☢☢

These imaging modalities were ordered by Neurosurgery

Findings (unlabeled)



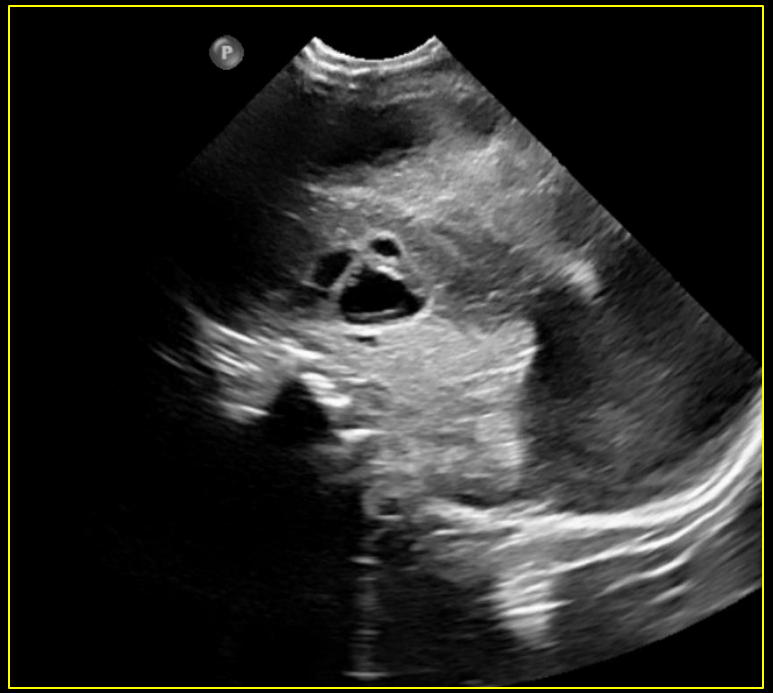
Axial T2W

Sagittal T2W DRIVE sequence

Findings (unlabeled)

Axial T2W

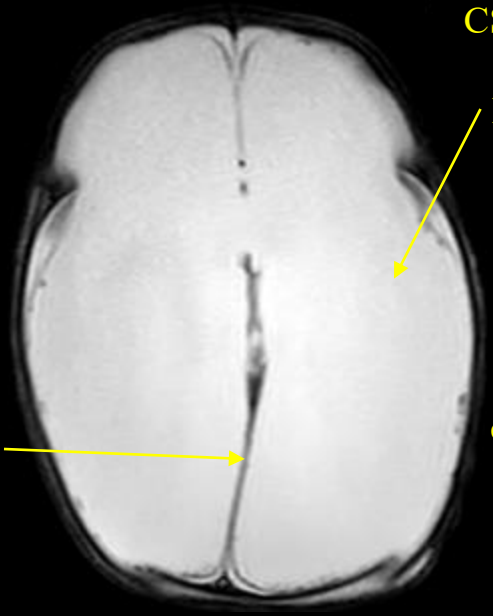
Findings (unlabeled)



Sagittal Head US

Coronal Head US

Findings (labeled)

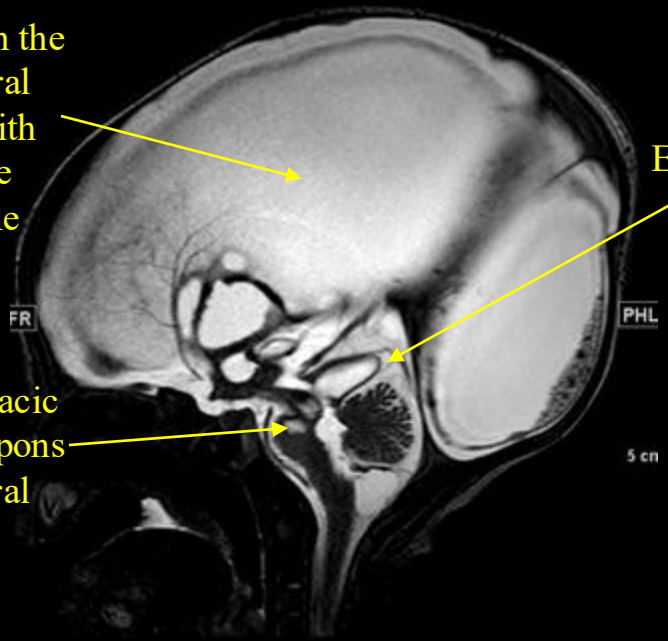


Presence of falx cerebri

Axial T2W

CSF filled sacs in the bilateral cerebral hemispheres with absence of the cerebral mantle

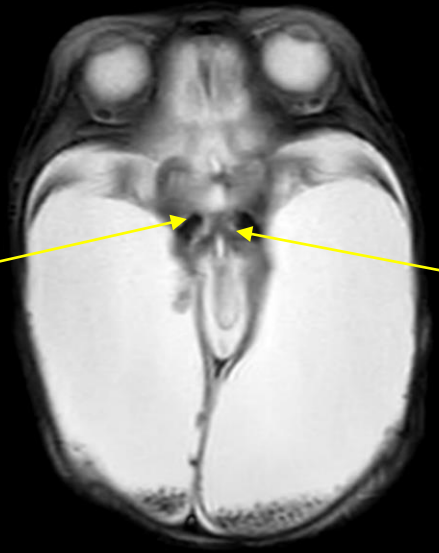
Encephalomalacic changes in the pons and the ventral midbrain



Encephalomalacic changes in the tectum

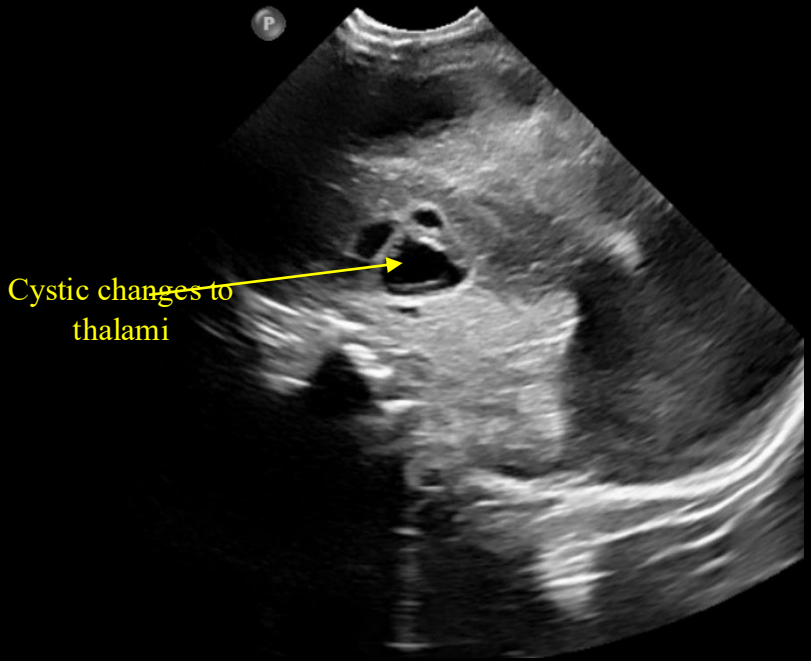
Sagittal T2W DRIVE sequence

Findings (labeled)

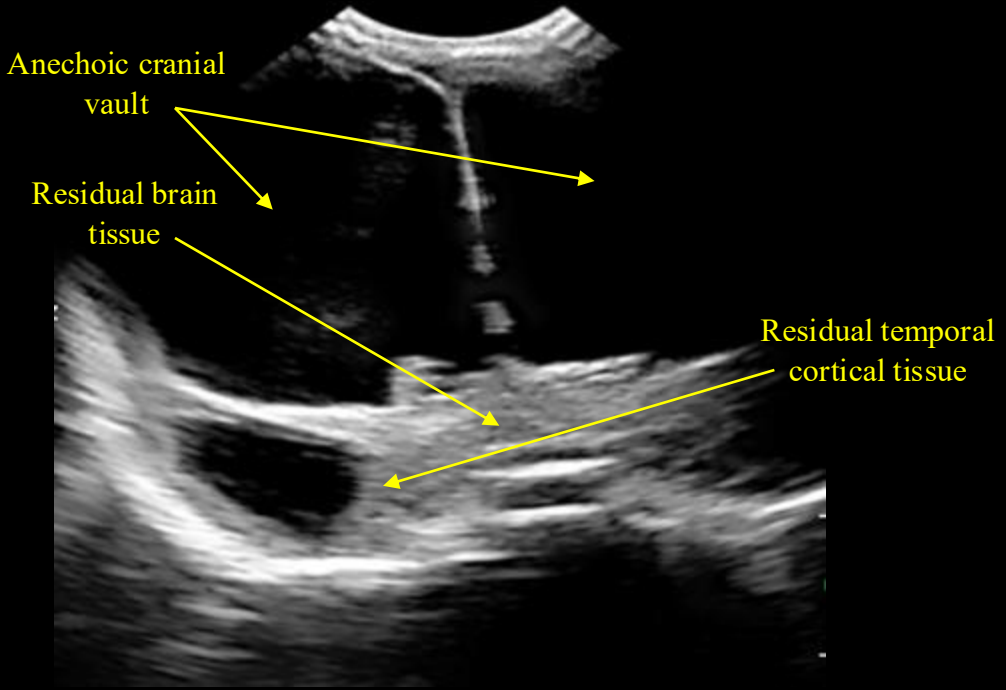


Axial T2W

Findings (labeled)



Sagittal Head US



Coronal Head US

Final Dx:

Hydranencephaly

Case Discussion: Differential Diagnosis

	Imaging features
Alobar prosencephaly (also known as lobar holoprosencephaly)[1,2]	-Absence of midline structures such as falx cerebri, corpus callosum, and septum pellucidum. -Fused basal ganglia/thalami -Midline facial abnormalities
Severe obstructive hydrocephalus[3]	-Aqueductal stenosis -Dilation of lateral and third ventricles -Identifiable (thinned) cortex -Preserved middle cerebral arteries (MCA) on vascular imaging
Hydranencephaly[4]	-Preserved thalami/posterior fossa -Presence of falx -Absence of MCA -Remanants of temporal/occipital lobes -Typically remaining brain structures have no gliosis
Severe Bilateral Open-Lip Schizencephaly[5]	-Cleft walls are open and filled with CSF -Cleft most often involves posterior frontal/parietal lobes -Frequently associated with other cerebral anomalies including grey

Case Discussion: Hydranencephaly+

- Hydranencephaly clinical presentation can include seizures, respiratory failure, flaccidity, or decerebrate posturing and a vegetative state.[6]
- Etiology - In utero compromise of the anterior circulation, leading to absent MCA and loss of cortical tissue. Likely maternal SUD is the contributing factor in our case, with vascular dysfunction and subsequent infarction of not only the bilateral cerebral hemispheres but also the deep gray nuclei and the brainstem.[7]
- Vasoconstrictive agents have been associated with hydranencephaly, and there are limited examples of cocaine specifically.[8, 9]
- Additional causes of hydranencephaly include intrauterine infection, fetal hypoxia (carbon monoxide exposure, genetic causes (mutations in COL4A1 gene, LAMB1 gene, and PI3K-Akt3-mTOR pathway), twin pregnancy, and rare syndromes including fowler syndrome, deficiency of factor XIII, and intracerebral hemorrhage.[10]

Case Discussion: Hydranencephaly+

Hydranencephaly	Hydranencephaly+
-Preserved thalami/posterior fossa -Presence of falx -Absence of MCA -Remanants of temporal/occipital lobes -Typically remaining brain structures have no gliosis	-Features of hydranencephaly present -Additional gliosis of residual cerebral tissue not supplied by MCA, including the brainstem and posterior fossa.

References

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