AMSER Case of the Month June 2026

5-day-old male born at 39 weeks with abnormal newborn screening positive for elevated leucine

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Patient Presentation

HPI

- 5-day-old male born at 39 weeks presented to the ED after abnormal newborn screen notable for markedly elevated leucine
- Mother reported upper extremity boxing movements at home
- Admitted to NICU; encephalopathic with subclinical seizures requiring treatment
- Course complicated by cerebral edema and progressive respiratory failure requiring intubation

Physical Exam

- General: Minimal spontaneous activity, not in distress but encephalopathic
- Neurologic: Encephalopathic; stirs to exam with intermittent shivering; movements of unclear intentionality

Pertinent Labs

- **Plasma amino acids:**
  - Leucine >4000 µmol/L (initial)
  - Isoleucine ↑ (~227–243 µmol/L)
  - Valine ↑ (~270–296 µmol/L)
- Acid–base: initial metabolic acidosis
- Electrolytes: recurrent hypokalemia (K ~2.3–3.1 mmol/L)
- Albumin: low (~2.0–2.2 g/dL)
- CBC: anemia (Hgb nadir ~9 g/dL), thrombocytopenia (~100–110K)
- Urinalysis: initial ketonuria (4+)

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

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

Procedure	Appropriateness Category	Peds Relative Radiation Level
MRI head without IV contrast	Usually Appropriate	0
US head	May Be Appropriate	0
MRI head without and with IV contrast	May Be Appropriate	0
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	☢☢☢☢
FDG-PET/CT brain	Usually Not Appropriate	☢☢☢☢
SPECT or SPECT/CT brain perfusion	Usually Not Appropriate	☢☢☢☢

These imaging modalities were ordered

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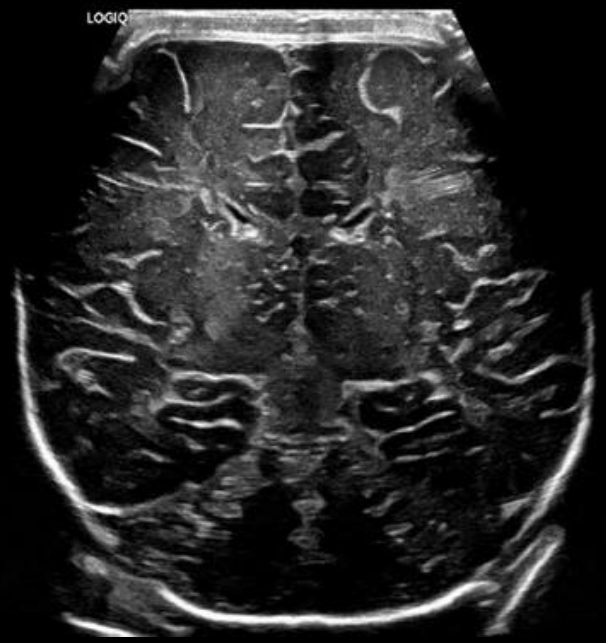
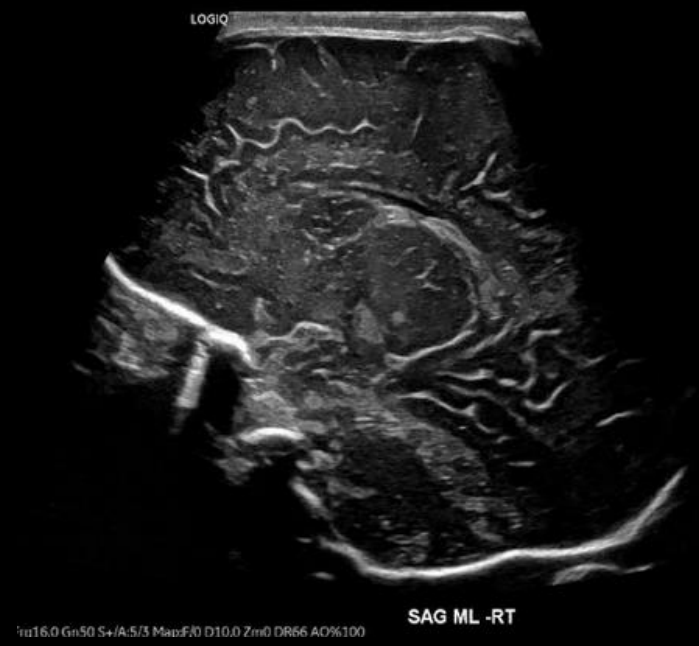
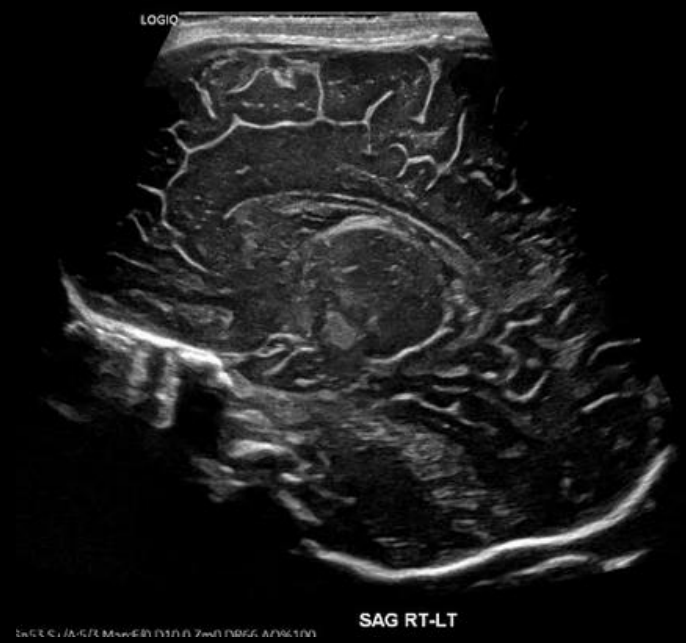
Findings (unlabeled)







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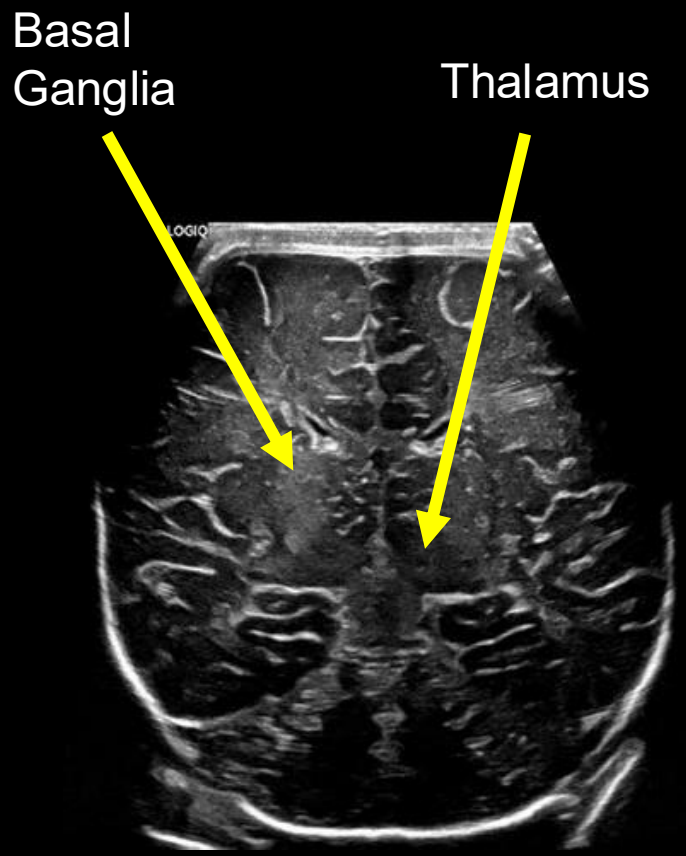
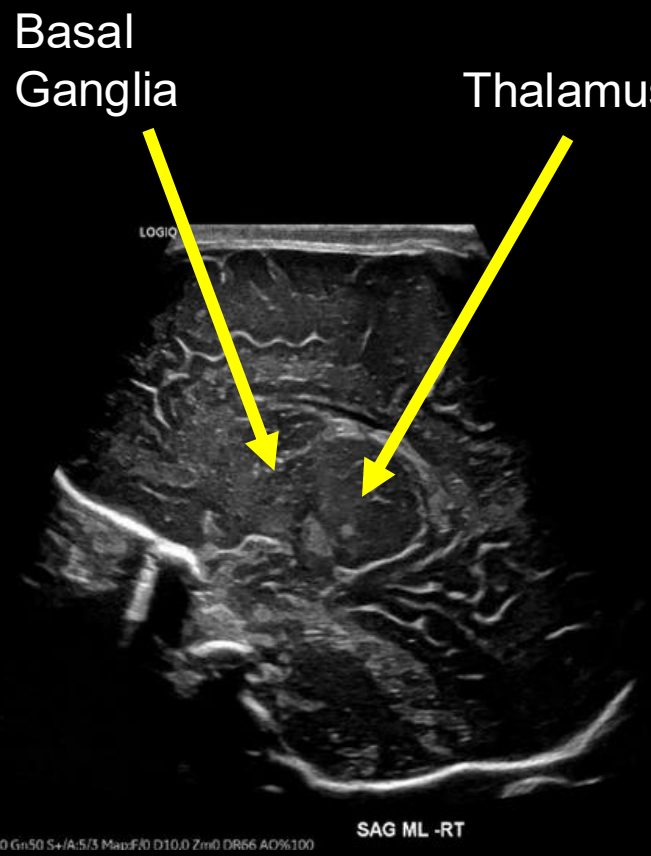
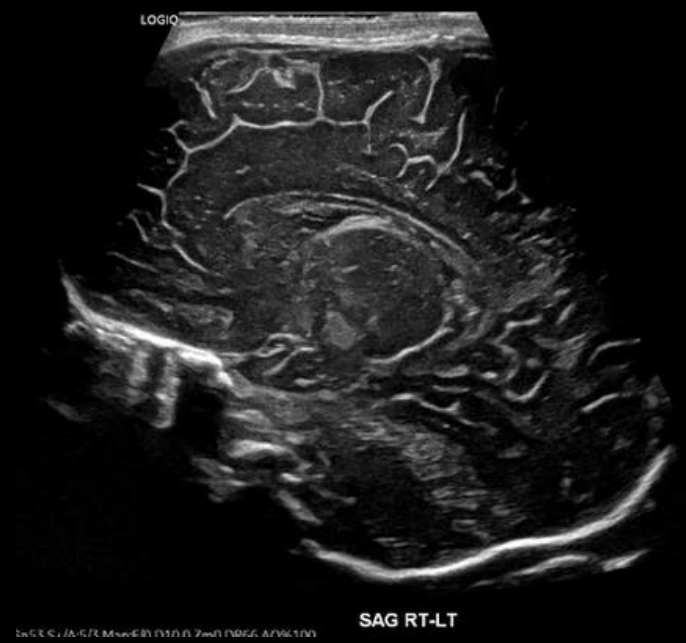


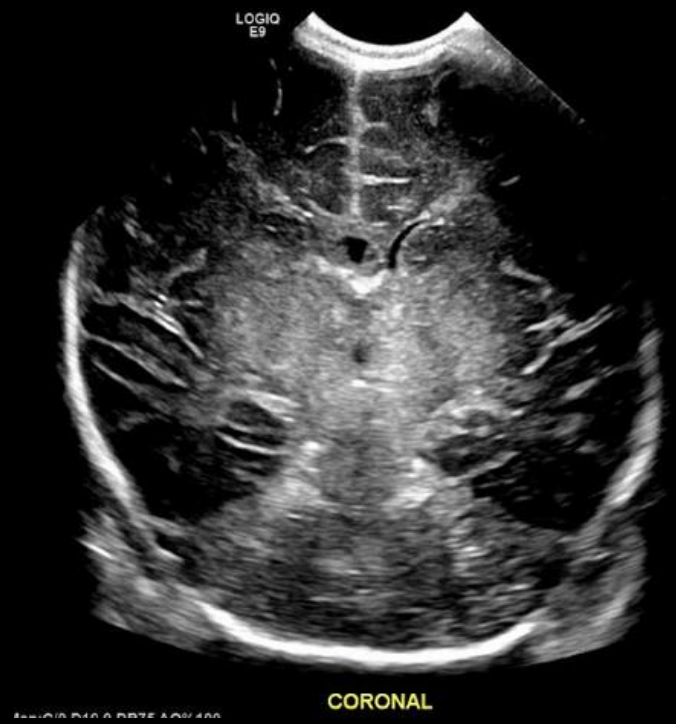


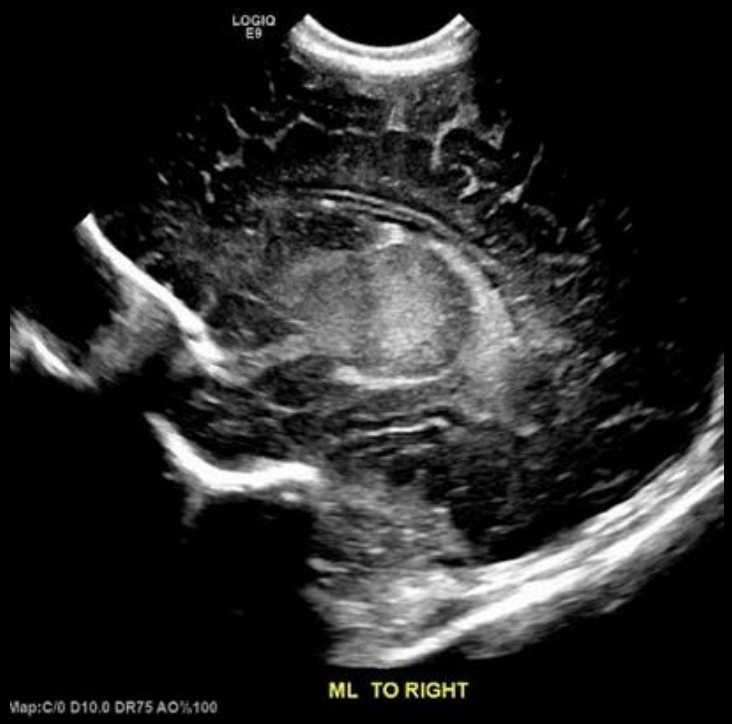


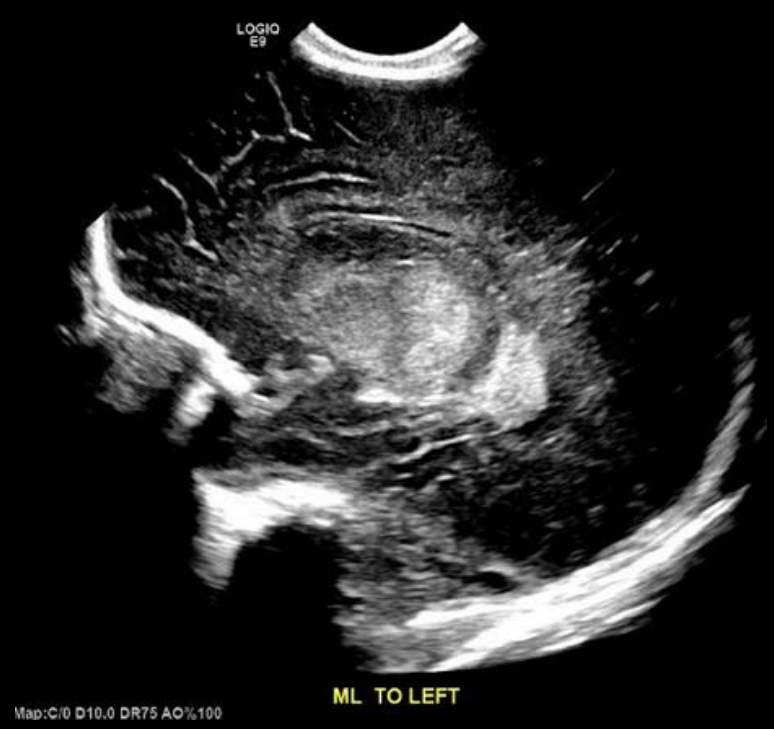
Normal head ultrasound with normal echogenicity of the parenchyma

Ultrasound was repeated 8 days later

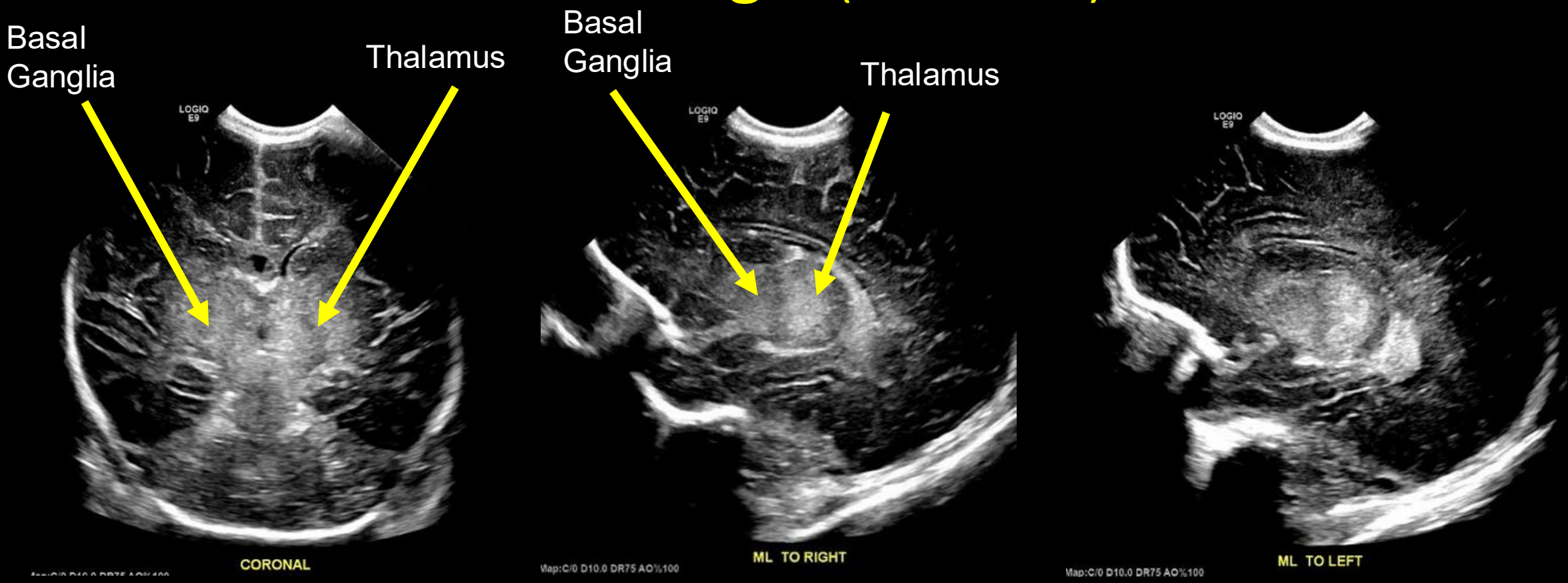
Findings: (unlabeled)







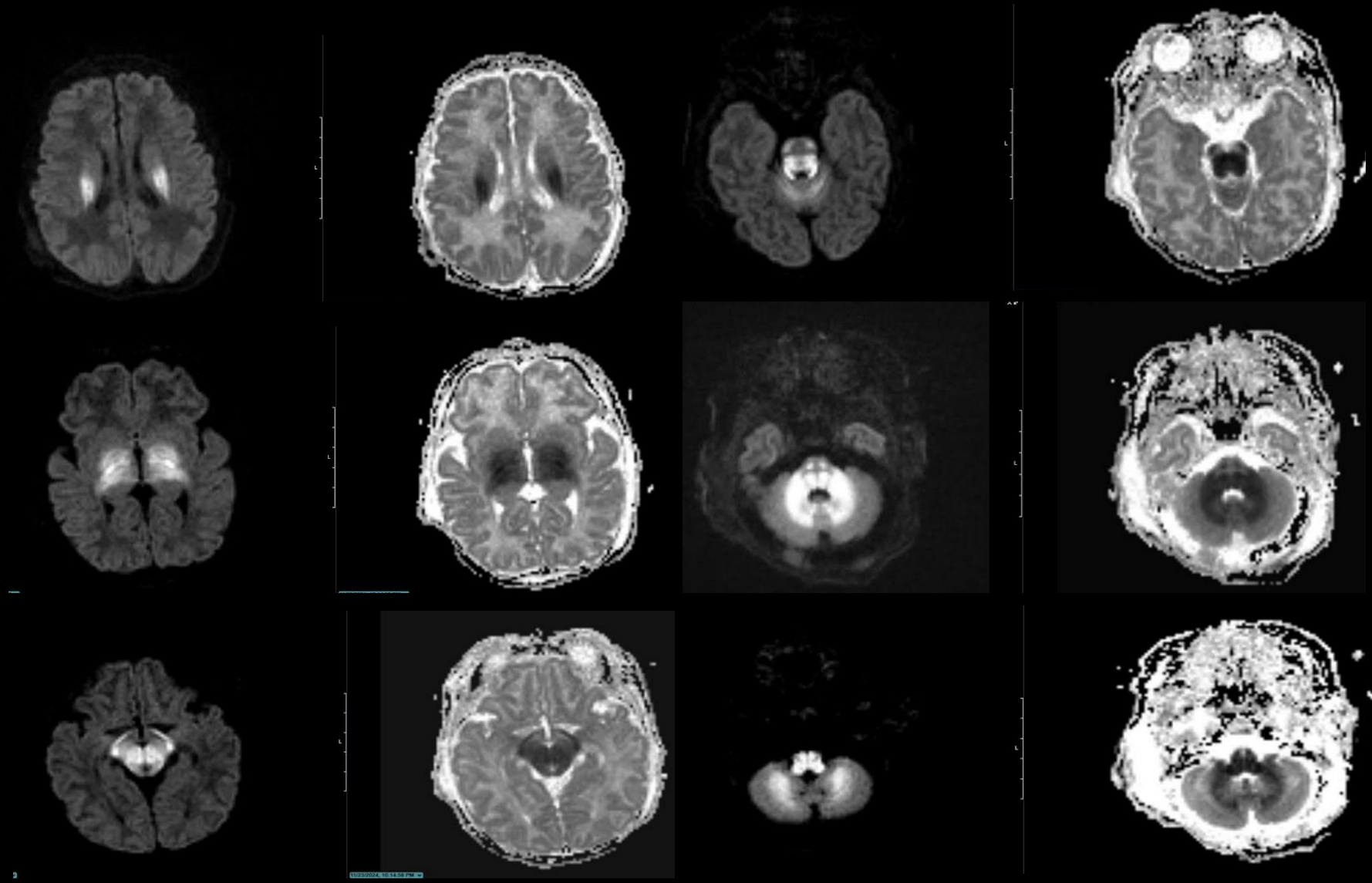
Findings: (labeled)



There is abnormally increased echogenicity in the basal ganglia/thalami consistent with developing cerebral edema

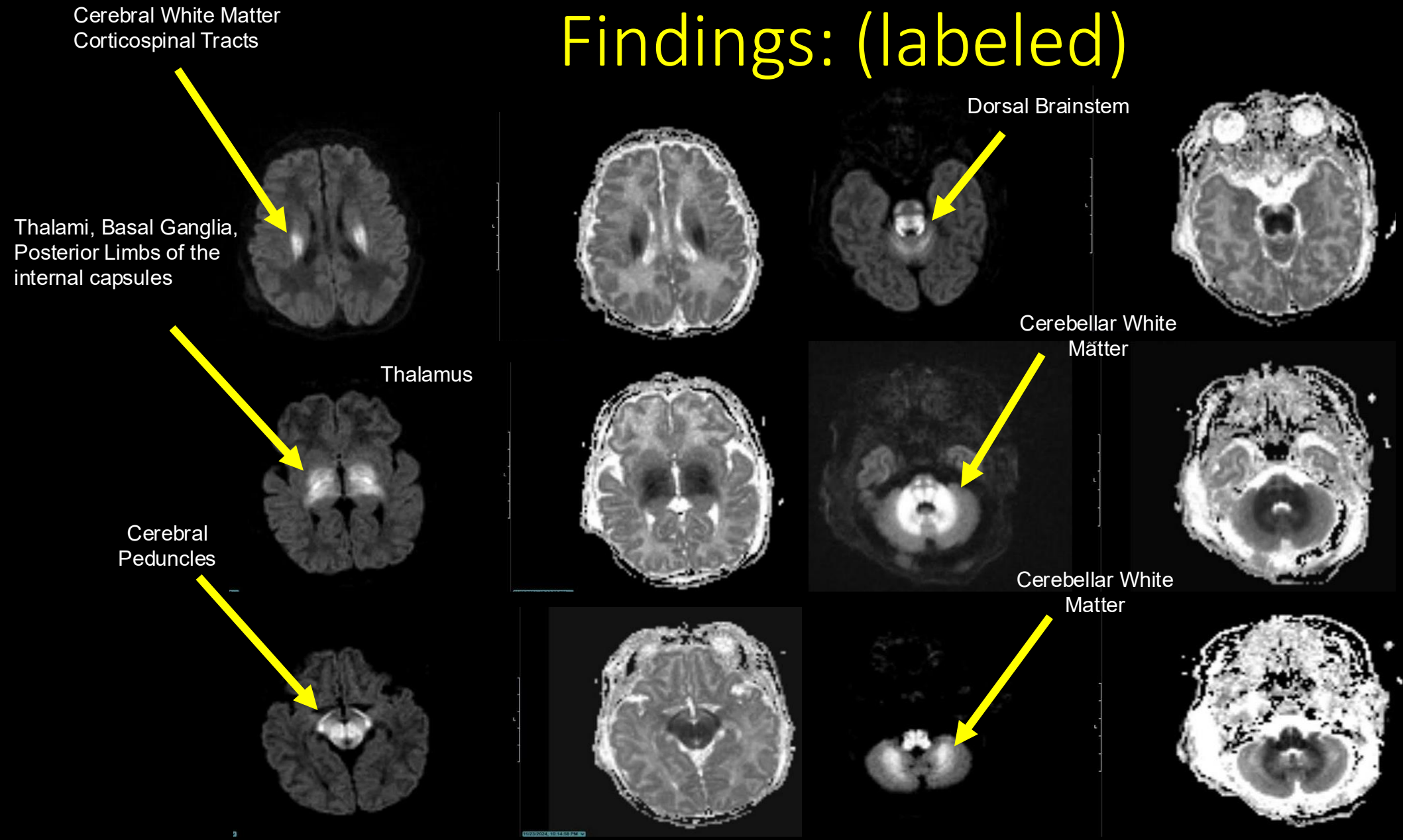
MRI was ordered to confirm findings

Findings: (unlabeled)



Abnormal diffusion restriction (bright signal on DWI sequence with corresponding dark signal on ADC map) in several areas including the cerebellar white matter, cerebral peduncles, dorsal brainstem, posterior limbs of the internal capsules, thalami, basal ganglia, and cerebral white matter corticospinal tracts. Findings are consistent with intramyelinic edema (potentially reversible).

Findings: (labeled)



Final Dx:
Maple Syrup Urine Disease

Case Discussion

- **Pathophysiology**
  - Autosomal recessive disorder of branched-chain amino acid (BCAA) metabolism[1,2,3]
  - Caused by deficiency of the branched-chain alpha-keto acid dehydrogenase complex[1,2,3]
  - Leucine accumulation → neurotoxicity via osmotic and excitotoxic mechanisms → cerebral edema[1,2,3]
  - Symptoms begin in the first week of life after protein feeding: poor feeding, lethargy, vomiting, abnormal tone/movements[1,2,3]

Case Discussion

- **Imaging in MSUD Crisis**
  - **Head Ultrasound:** May show increased echogenicity in basal ganglia and thalami during crisis, reflecting intramyelinic edema (potentially reversible).
  - **MRI DWI:** Most sensitive modality; demonstrates restricted diffusion in a characteristic distribution[1,3,4]
    - Cerebellar white matter, cerebral peduncles, dorsal brainstem[1,3,4]
    - Posterior limbs of internal capsules, thalami, basal ganglia, corticospinal tracts[1,3,4]
  - Pattern mirrors active myelination at the time of metabolic crisis[1,3,4]

Case Discussion

- **Treatment**
  - **Acute Management:**
    - Dietary BCAA restriction[2]
    - High-calorie supplementation to suppress catabolism[2]
    - In severe cases: continuous venovenous hemofiltration (CVVH) to rapidly reduce leucine levels[2]
  - **Long-term Management:**
    - Strict low-BCAA diet with metabolic formula supplementation[2]
    - Liver transplantation is curative[2]

References:

1. Kilicarslan R, Alkan A, Demirkol D, Toprak H, Sharifov R. Maple syrup urine disease: diffusion-weighted MRI findings during acute metabolic encephalopathic crisis. *Jpn J Radiol.* 2012;30(6):522-525. doi:10.1007/s11604-012-0079-2.

2. Scharre S, Mengler K, Schnabel E, et al. Impact of early diagnosis, disease variant, and quality of care on the neurocognitive outcome in maple syrup urine disease: A meta-analysis. *Genet Med.* 2025;27(1):101303. doi:10.1016/j.gim.2024.101303.

3. Liu Q, Li F, Zhou J, Liu X, Peng J, Gong L. Neonatal maple syrup urine disease case report and literature review. *Medicine (Baltimore).* 2022;101(50):e32174. doi:10.1097/MD.0000000000032174.

4. Li Y, Liu X, Duan CF, Song XF, Zhuang XH. Brain magnetic resonance imaging findings and radiologic review of maple syrup urine disease: Report of three cases. *World J Clin Cases.* 2021;9(8):1844-1852. doi:10.12998/wjcc.v9.i8.1844.