AMSER Case of the Month
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Four-month-old male presents with new onset nonfebrile seizures

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Patient Presentation

• **HPI:** A previously healthy 16-week-old male presented with 2 seizure-like episodes in the setting of URI symptoms without reported fever. Episodes consisted of curled up hands and feet, full body stiffening and jolting movements, as well as non-responsiveness.

• **Developmental history:** Meeting milestones through 4 months

• **Vital signs:** BP 127/64, Pulse 147, Temp 99.4F, Resp 48, SpO2 99%

• **Physical Exam:** No acute distress, non-toxic appearing. Head lag, poor tracking. Salmon patch on forehead.
Pertinent Labs

- **CBC:** 4.65 WBC, 11.5 Hgb (WNL)
- **Lactate:** 2.1 (H)
- **Procalcitonin:** 0.19 (H)
- **Coronavirus SARS CoV 2:** Positive
- **LP:** Negative CSF infectious panel
What Imaging Should We Order?
Select the applicable ACR Appropriateness Criteria

**Variant 7:** Children 1 month to 17 years of age. Generalized seizure (neurologically abnormal). Initial imaging.

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<th>Procedure</th>
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<th>Relative Radiation Level</th>
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This imaging modality was ordered by the ER physician.
Findings (unlabeled)
Asymmetric increased size of the right frontal lobe with:

- Lobar cortical thickening
- Asymmetric enlargement of the right lateral ventricle
Should we pursue further imaging?
Select the applicable ACR Appropriateness Criteria

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Variant 7: Children 1 month to 17 years of age. Generalized seizure (neurologically abnormal). Initial imaging.

This imaging modality was ordered at discharge.
Findings (unlabeled)
Asymmetric increased size of the right frontal lobe with:

- Lobar cortical thickening
- Decreased sulcation and pachygyria
- Increased white matter volume
- Asymmetric enlargement of the right lateral ventricle
Differential Diagnosis

- Neuronal migration anomaly
  - Focal cortical dysplasia
  - Lissencephaly-pachygyria spectrum

- Enlarged unilateral hemisphere
  - Gliomatosis cerebri
  - Hemimegalencephaly

- Small unilateral hemisphere
  - Rasmussen encephalitis
  - Sturge-weber syndrome
  - Dyke-Davidoff-Masson syndrome
Final Dx:

Hemimegalencephaly

(Partial involving the right frontal lobe)
Hemimegalencephaly

- **Definition**: A rare cortical malformation disorder characterized by hamartomatous enlargement of part or entire cerebral hemisphere.

- **Etiology**: Abnormal activation of mTOR signaling pathway
  - Sporadic
  - Syndromic: Neurocutaneous syndromes, overgrowth syndromes with vascular malformations

- **Clinical features**:
  - Often diagnosed in the first year of life
  - Presents with seizures, often refractory to antiepileptic drugs; developmental delay; motor deficits; macrocrania
Hemimegalencephaly

• Imaging findings:
  • Ultrasound, CT, or MRI can help identify key imaging features
    • Enlarged cerebral lobe or hemisphere, enlarged ipsilateral lateral ventricle with pointed frontal horn, thickened cortex, dystrophic calcification
  • MRI best characterizes
    • Abnormal gray matter-white matter differentiation
    • Pachygyria, polymicrogyria, or gray matter heterotopia
    • Increased white matter volume, with decreased T2 signal intensity
  • SPECT/PET
    • Hypometabolism in the affected hemisphere
Hemimegalencephaly

• **Treatment:**
  - Medical management
    - Antiepileptic therapies often trialed but typically ineffective
  - Surgical management
    - Functional versus anatomic hemispherectomy
    - More recently, endovascular embolic hemispherectomy has been shown to be effective
    - Hemispherotomy results in seizure freedom rates of 50-90% with benefits in psychomotor outcomes associated with earlier intervention in patients with refractory seizures
References:


