AMSER Case of the Month August 2023

Four-month-old male presents with new onset nonfebrile seizures

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LOMA LINDA UNIVERSITY HEALTH

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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.**

Procedure	Appropriateness Category	Relative Radiation Level
MRI head without IV contrast	Usually Appropriate	O
MRI head without and with IV contrast	May Be Appropriate	O
CT head without IV contrast	May Be Appropriate	☢☢☢
US head	Usually Not Appropriate	O
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	☢☢☢☢

This imaging modality was ordered by the ER physician

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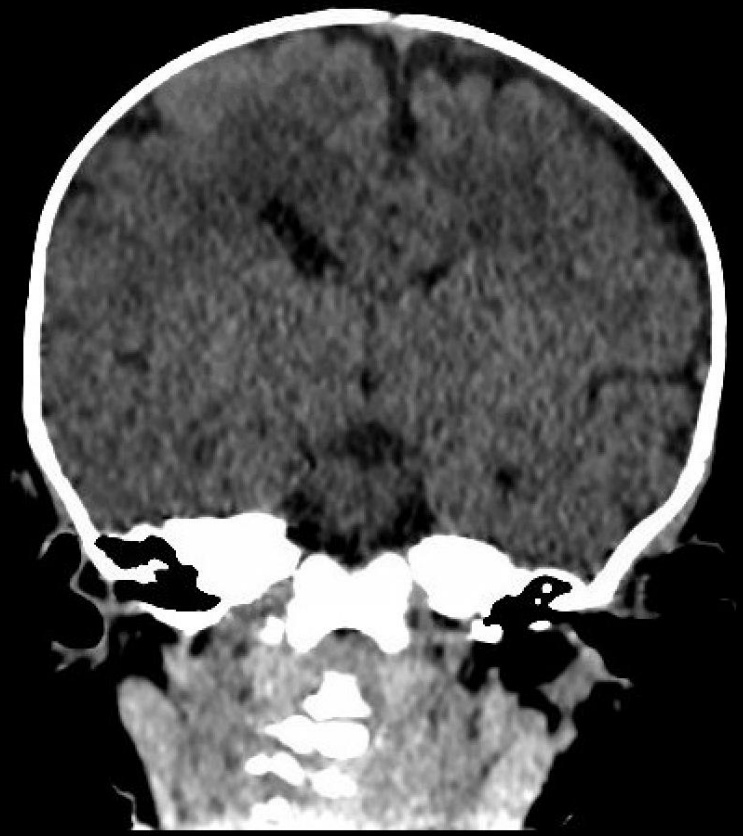
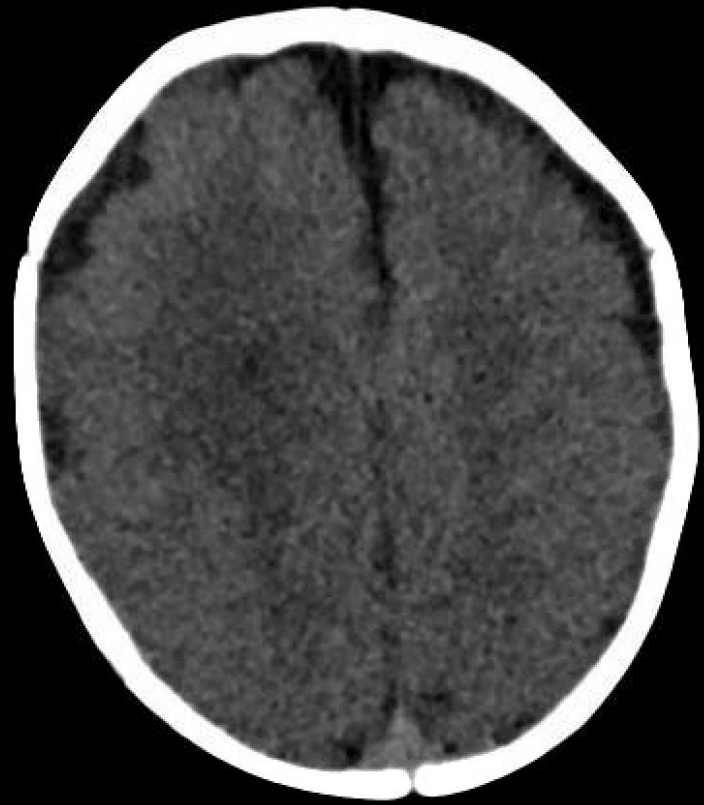
Findings (unlabeled)

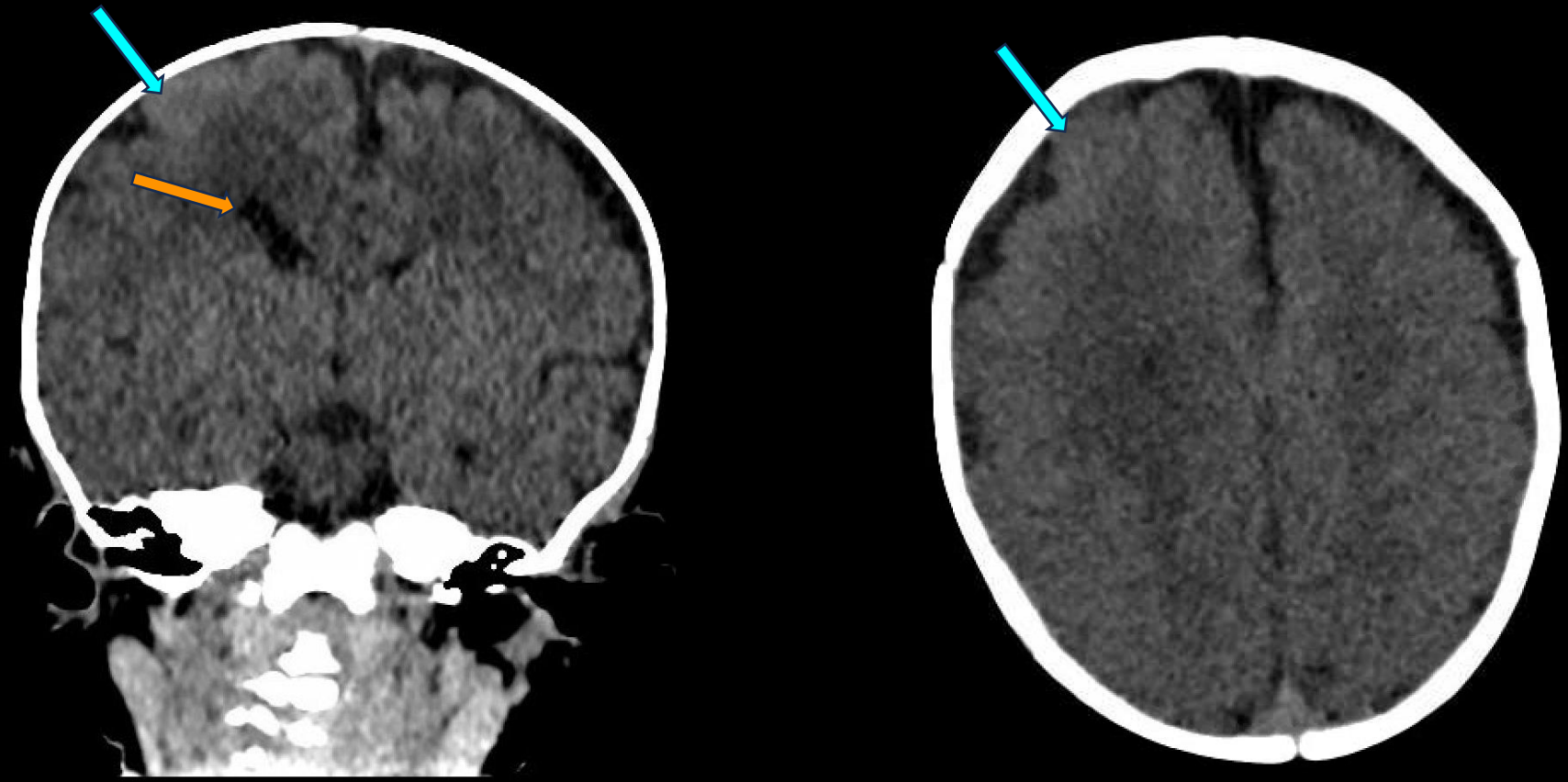




Asymmetric increased size of the right frontal lobe with:

- Lobar cortical thickening
- Asymmetric enlargement of the right lateral ventricle

Findings: (labeled)



Should we pursue further imaging?

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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MRI head without IV contrast	Usually Appropriate	O
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HMPAO SPECT or SPECT/CT brain	Usually Not Appropriate	☢☢☢☢
FDG-PET/CT brain	Usually Not Appropriate	☢☢☢☢

This imaging modality was ordered at discharge

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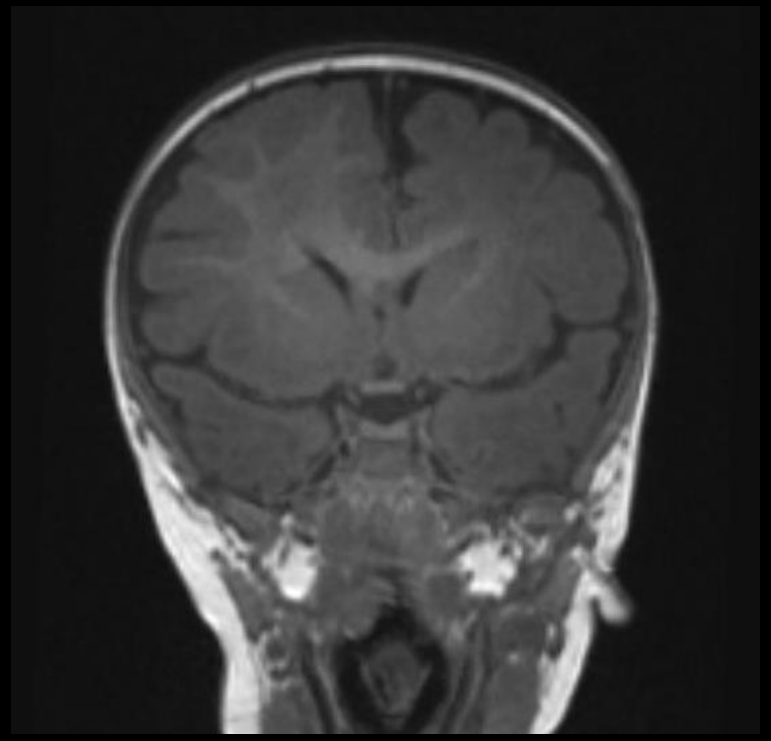
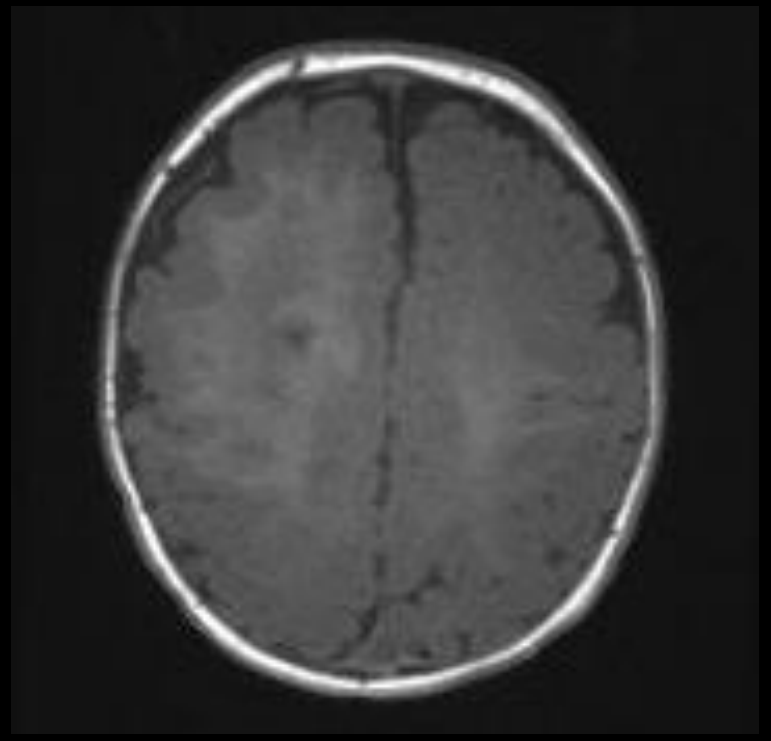
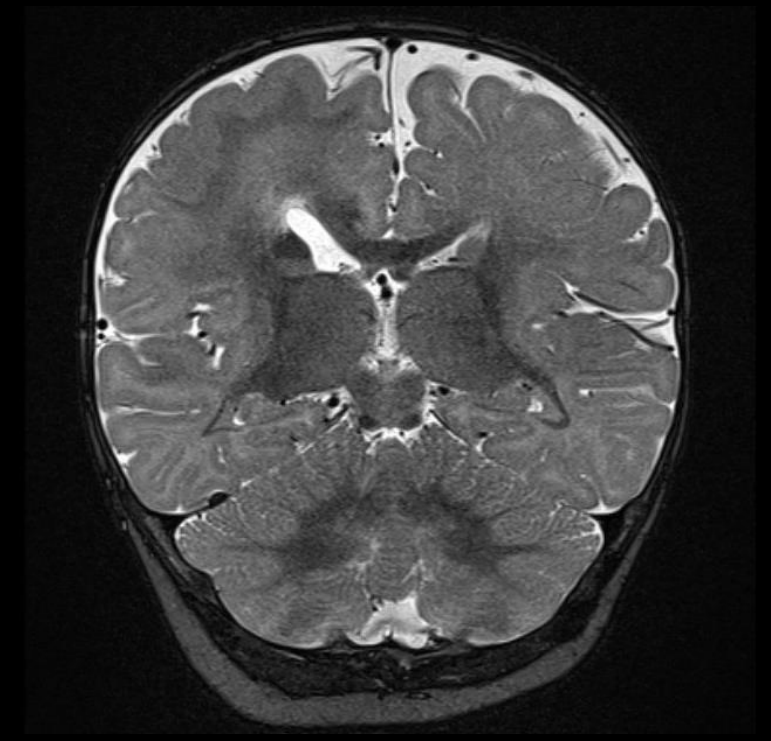
Findings (unlabeled)



Coronal T1



Axial T1

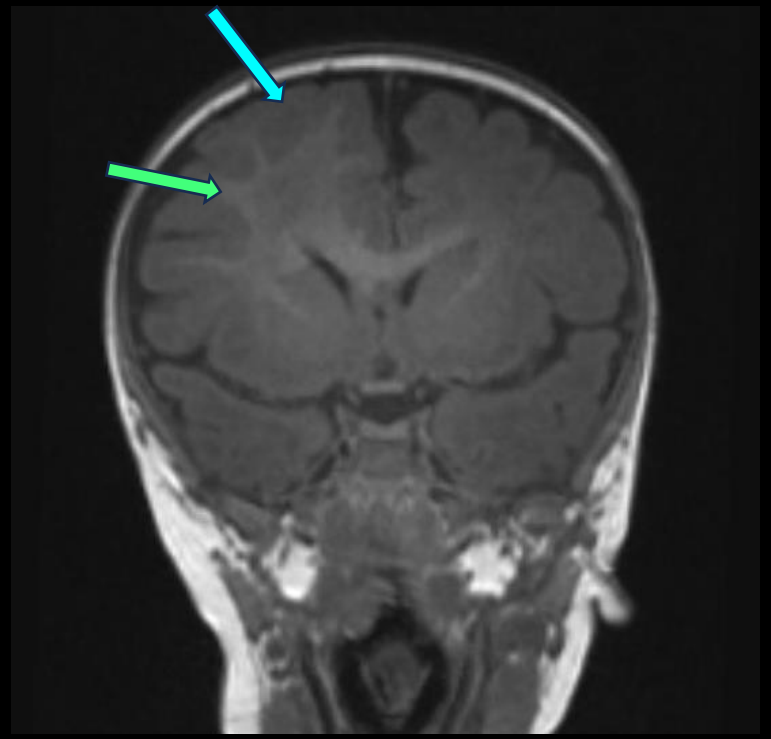


Coronal T2

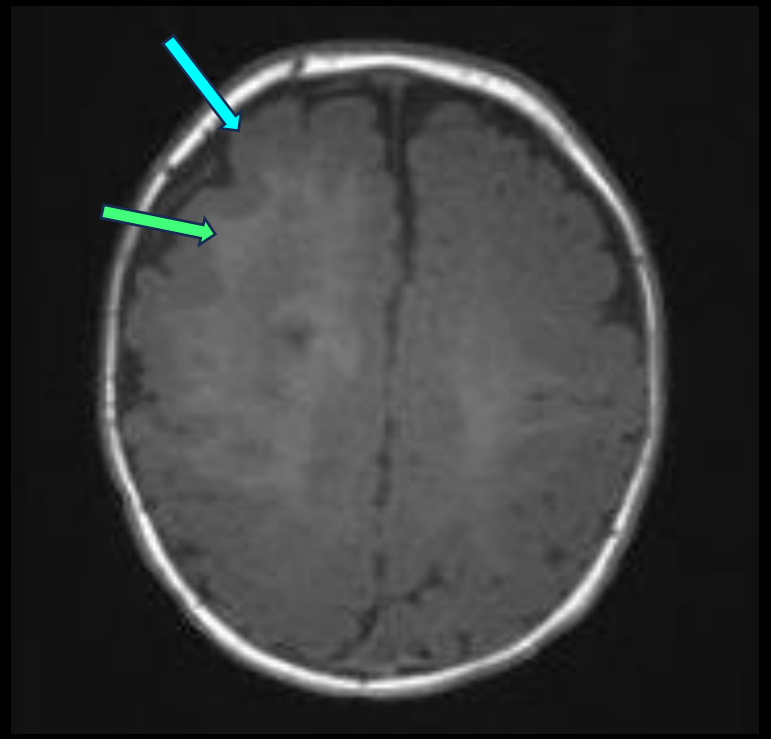
Asymmetric increased size of the right frontal lobe with:

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

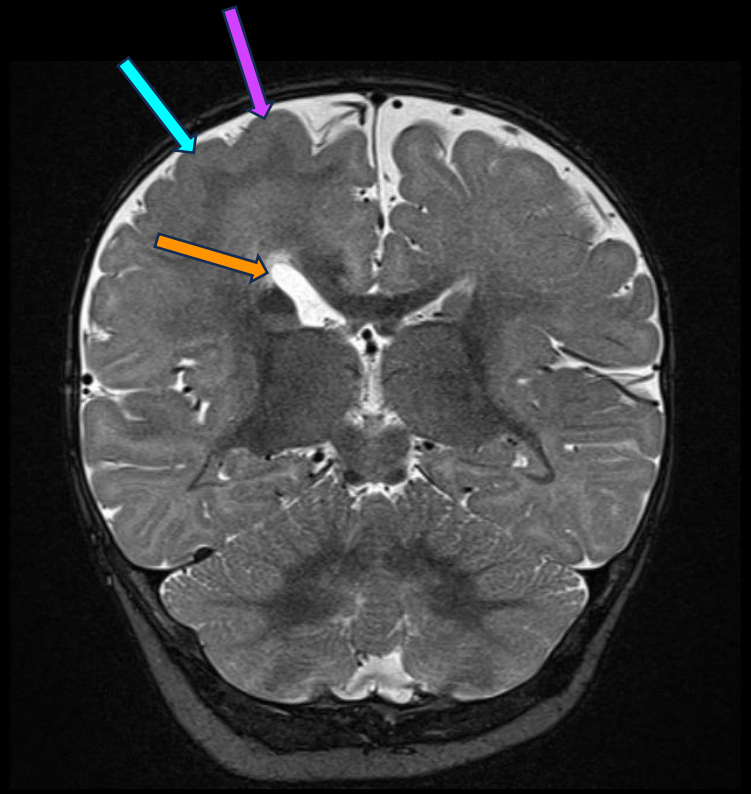
Findings (labeled)



Coronal T1



Axial T1



Coronal T2

Differential Diagnosis

- Neuronal migration anomaly
  - Focal cortical dysplasia
  - Lissencephaly-pachygyria spectrum
- Enlarged unilateral hemisphere
  - Gliomatosis cerebri
  - Hemimegalencephaly
- **S**mall 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:

Di Rocco C, Battaglia D, Pietrini D, Piastra M, Massimi L. Hemimegalencephaly: clinical implications and surgical treatment. *Childs Nerv Syst*. 2006;22(8):852-866.

Garcia CAB, Carvalho SCS, Yang X, et al. mTOR pathway somatic variants and the molecular pathogenesis of hemimegalencephaly. *Epilepsia Open*. 2020;5(1):97-106.

Jaiswal V, Hanif M, Sarfraz Z, et al. Hemimegalencephaly: A rare congenital malformation of cortical development. *Clin Case Rep*. 2021;9(12):e05238.

Pepi C, De Benedictis A, Rossi-Espagnet MC, et al. Hemispherotomy in infants with hemimegalencephaly: Long-term seizure and developmental outcome in early treated patients. *Brain Sci*. 2022;13(1):73.

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