AMSER Case of the Month August 2025

6-year-old female with history of developmental delay presents with reported seizures

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Patient Presentation

- Mother of patient reports that starting in late 2022, patient began having episodes of blank staring and unresponsiveness without shaking or loss of consciousness.
  - These episodes last 30 seconds – 1 minute and the patient is tired afterwards
- Occasionally associated with these episodes is tachycardia (120s-140s at rest) and hypertension for which she is following with Pediatric Cardiology

Patient Presentation

- Mom reports that patient has a history of delayed developmental milestones, as well as abnormal gait and bilateral hand tremors
- Additionally, patient was fully continent but has recently occasionally been having episodes of bowel and bladder incontinence, not definitively related to the possible seizure events
- Has undergone two prior EEGs with some possible abnormalities but without clear epileptiform discharges
- Patient is scheduled for the EMU to be evaluated further by neurology

Pertinent Labs

- TSH, HbA1C, BMP all WNL
- No other pertinent labs

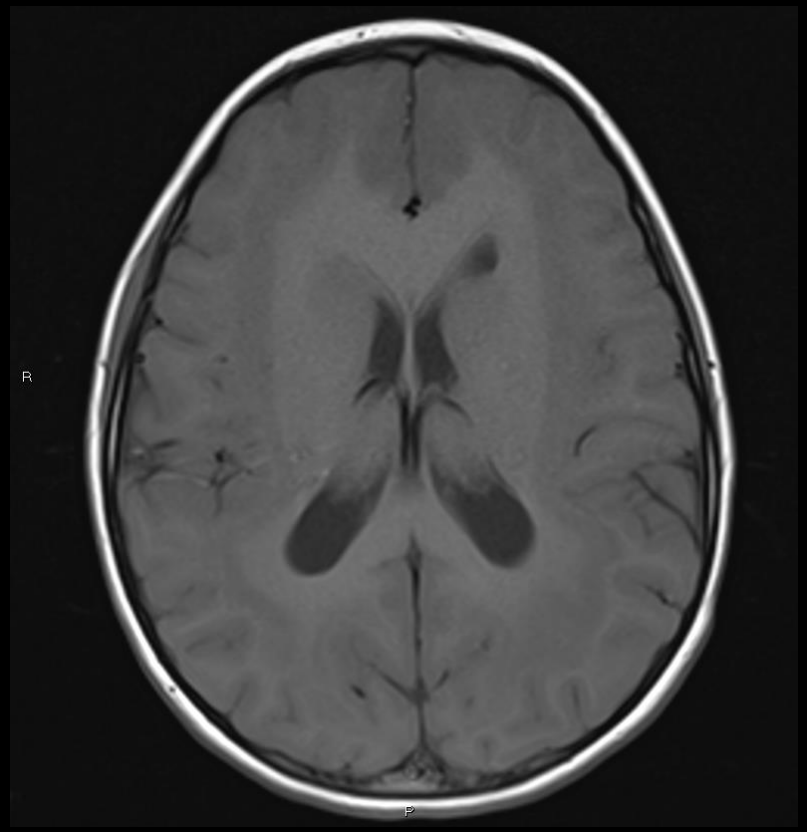
What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

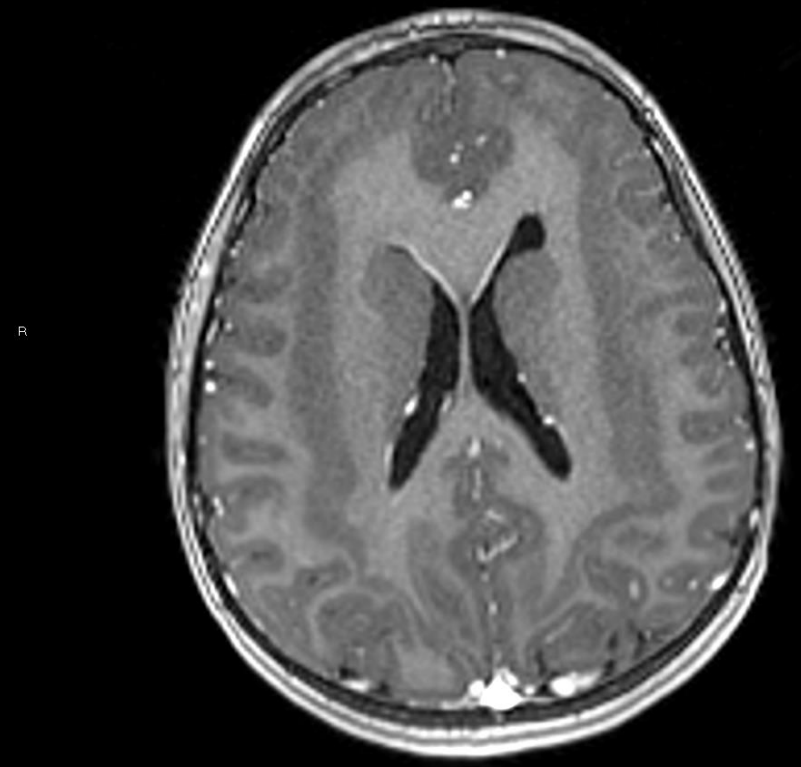
Scenario	Scenario ID	Procedure	Adult RRL	Peds RRL	Appropriateness Category
Seizure, generalized, neuro deficit, initial imaging	3074062	MRI head without IV contrast	0 mSv O	0 mSv [ped] O	Usually appropriate
		MRI head without and with IV contrast	0 mSv O	0 mSv [ped] O	May be appropriate
		CT head without IV contrast	1-10 mSv ☢☢☢	0.3-3 mSv [ped] ☢☢☢	May be appropriate
		US head	0 mSv O	0 mSv [ped] O	Usually not appropriate
		CT head with IV contrast	1-10 mSv ☢☢☢	0.3-3 mSv [ped] ☢☢☢	Usually not appropriate
		CT head without and with IV contrast	1-10 mSv ☢☢☢	3-10 mSv [ped] ☢☢☢☢	Usually not appropriate
		FDG-PET/CT brain	1-10 mSv ☢☢☢	3-10 mSv [ped] ☢☢☢☢	Usually not appropriate
		SPECT or SPECT/CT brain perfusion	1-10 mSv ☢☢☢	3-10 mSv [ped] ☢☢☢☢	Usually not appropriate

This imaging modality was ordered by the physician

Findings (unlabeled)

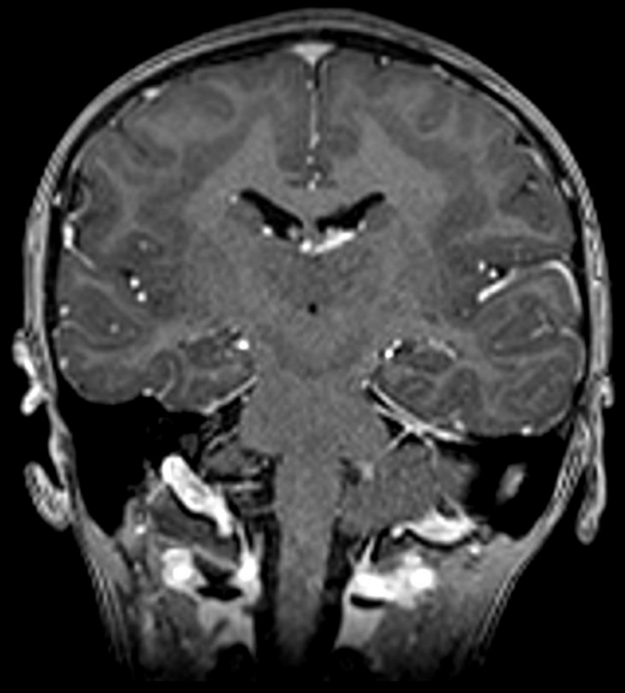


MRI T1 Axial



MPRAGE
Post-
Contrast Axial

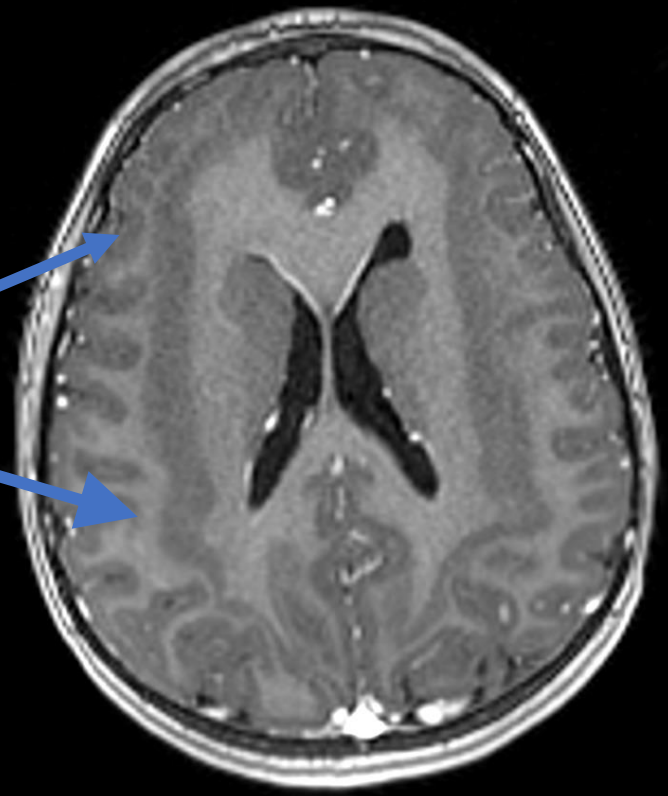
Findings (unlabeled)



MRI MPRAGE
Post-Contrast Coronal

Findings: (labeled)

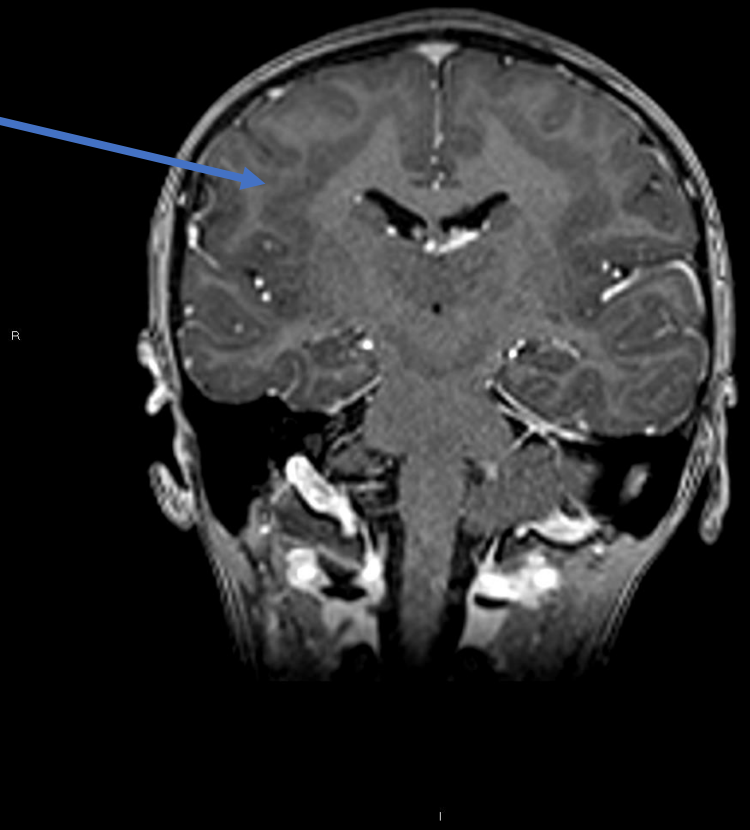
Extensive, thick band of gray matter heterotopia involving both the anterior and posterior aspects of the cerebral hemispheres



No evidence of pachygyria

Findings: (labeled)

Single continuous band of heterotopic gray matter outlining the underlying white matter structures



Final Dx:

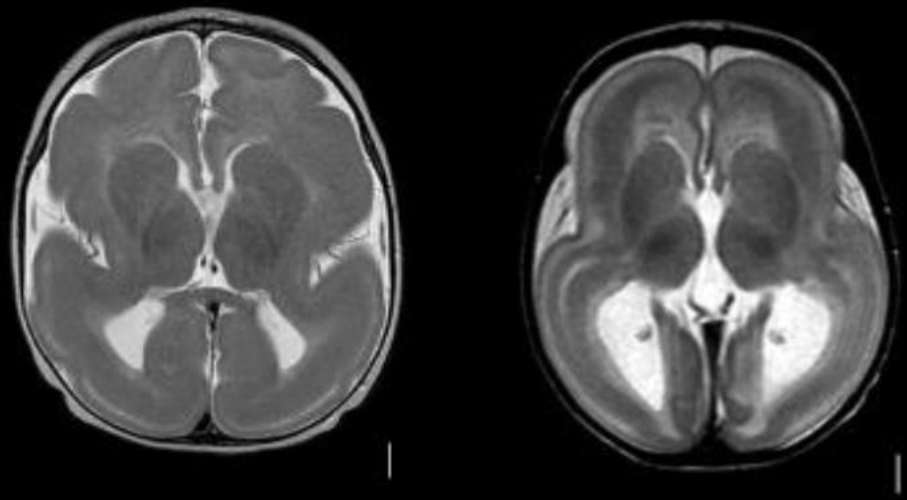
Band Heterotopia (Double Cortex Syndrome)

DCX mutation

Case Discussion

- Developmental delay and epilepsy may be seen in malformations of cortical development which occur due to abnormal neuronal migration during development
- Subcortical band heterotopia ("double cortex" syndrome) is an X-linked disorder of neuronal migration, presenting almost exclusively in females[1,3]
- The DCX gene mutation (which encodes a microtubule-associated protein) is the causative factor in most cases[2] accounting for 80% of cases of subcortical band heterotopias[3]
- DCX-related disorders in males presents as classic lissencephaly[4]

Di Donato N, Chiari S, Mirzaa GM, et al.



Lissencephaly
Decreased sulci and gyri (pachygyria, agyria)

Case Discussion

- Neuroimaging is essential for diagnosis, with MRI revealing continuous or diffuse band of grey matter below the cortex
- The imaging findings (thickness of grey matter band, presence of pachygyria, ventriculomegaly) correlate with the severity of the clinical presentation[5]
- Clinical presentation of subcortical heterotopia in females ranges from normal intelligence to mild or moderate intellectual disability. Epilepsy is common, with focal or generalized seizures[5]
- Agyria or pachygyria may also be present

Management

- Management is symptomatic and supportive for cognitive and developmental deficits
- Anti-epileptics are commonly used for symptom management [6]

The patient received genetic testing with confirmed DCX mutation.

She follows with Pediatric Neurology and is being treated with Keppra.

References

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- Haverfield EV, Whited AJ, Petras KS, Dobyns WB, Das S. Intragenic deletions and duplications of the LIS1 and DCX genes: a major disease-causing mechanism in lissencephaly and subcortical band heterotopia. *Eur J Hum Genet*. 2009;17(7):911-918. doi:10.1038/ejhg.2008.213
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