AMSER Case of the Month July 2026

12-year-old male with developmental delay presenting with new-onset seizures

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Patient Presentation

HPI: A 12-year-old male with a history of global developmental delay presented to pediatric neurology for evaluation of new-onset seizure activity.

PMH: The patient was born full-term in Africa without reported perinatal complications. He has a history of significant developmental delay. He began speaking at approximately 9–10 years of age but remains unable to sustain conversation, typically responding only with brief answers. He has also been non-ambulatory since birth.

Pertinent Labs and Physical Exam

- Labs: N/A
- Physical Exam: The patient demonstrated diffuse muscle atrophy and truncal hypotonia. Consistent with the reported history, the patient is unable to ambulate independently.
- EEG: Diffuse background slowing, consistent with mild generalized cerebral dysfunction.

What Imaging Should We Order?

ACR Appropriateness Criteria

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

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 pediatric neurologist.

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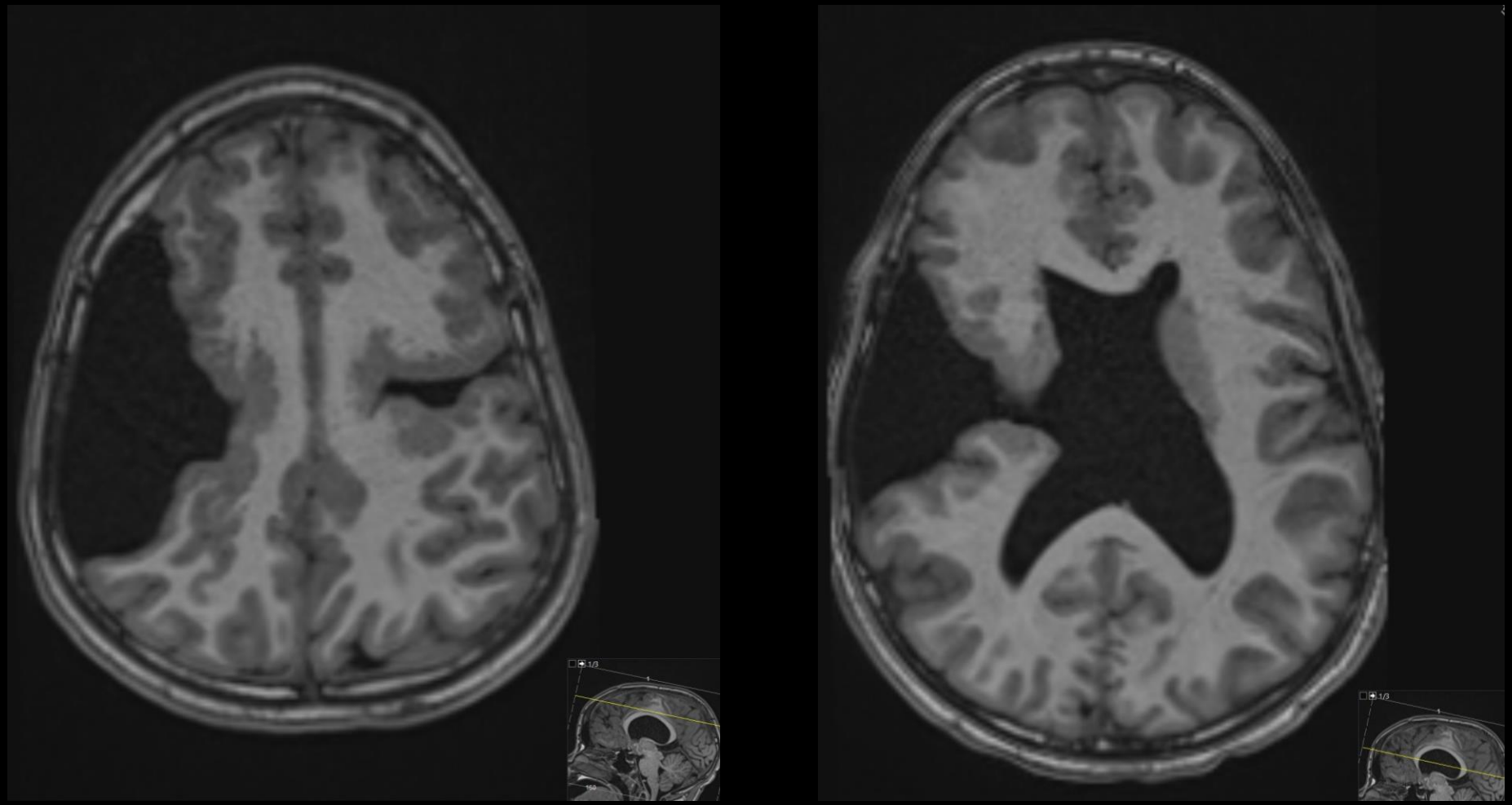
Findings (unlabeled)



Axial T1 MPRAGE MRI Sequence

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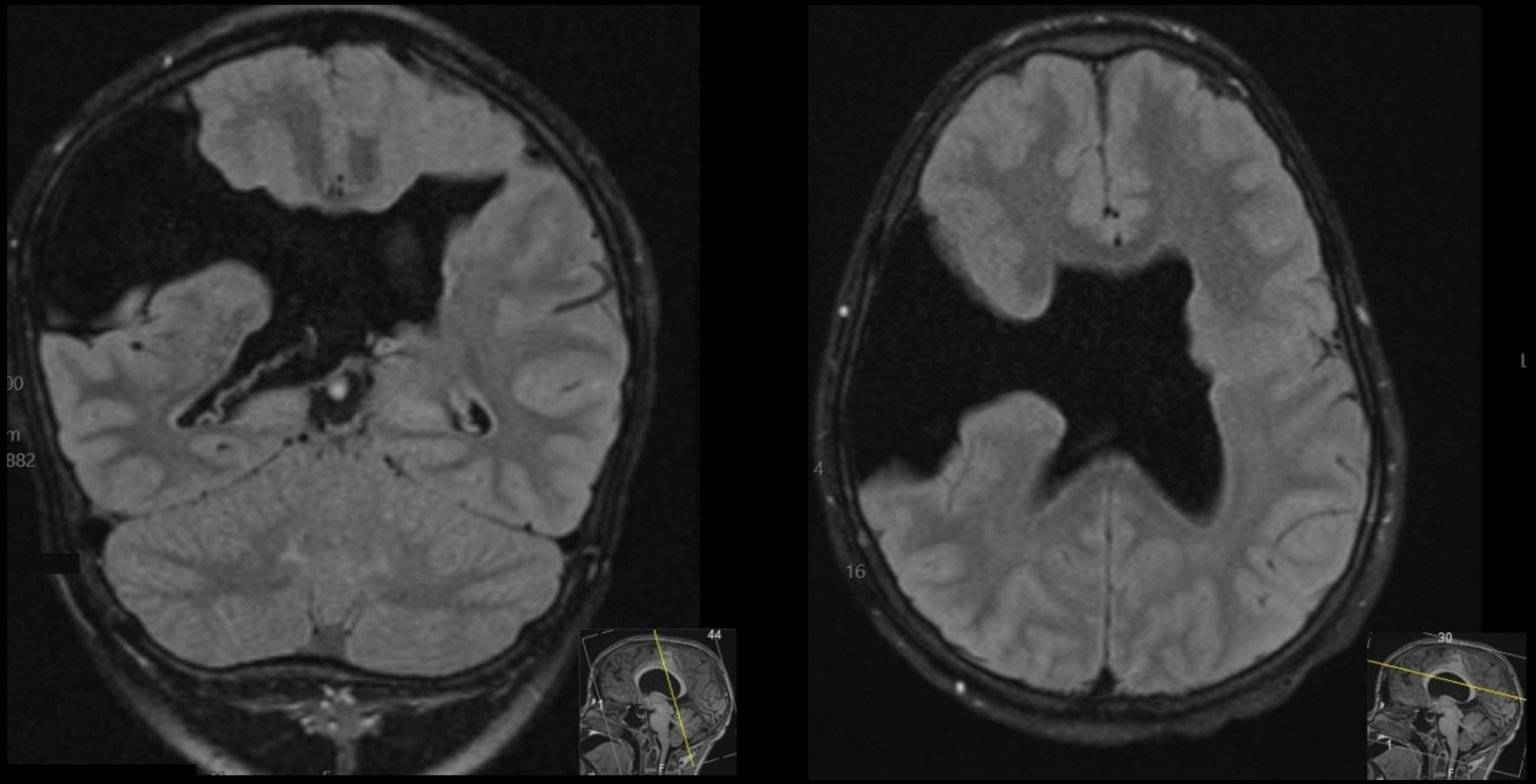
Findings (unlabeled)



Coronal and Axial FLAIR MRI Sequences

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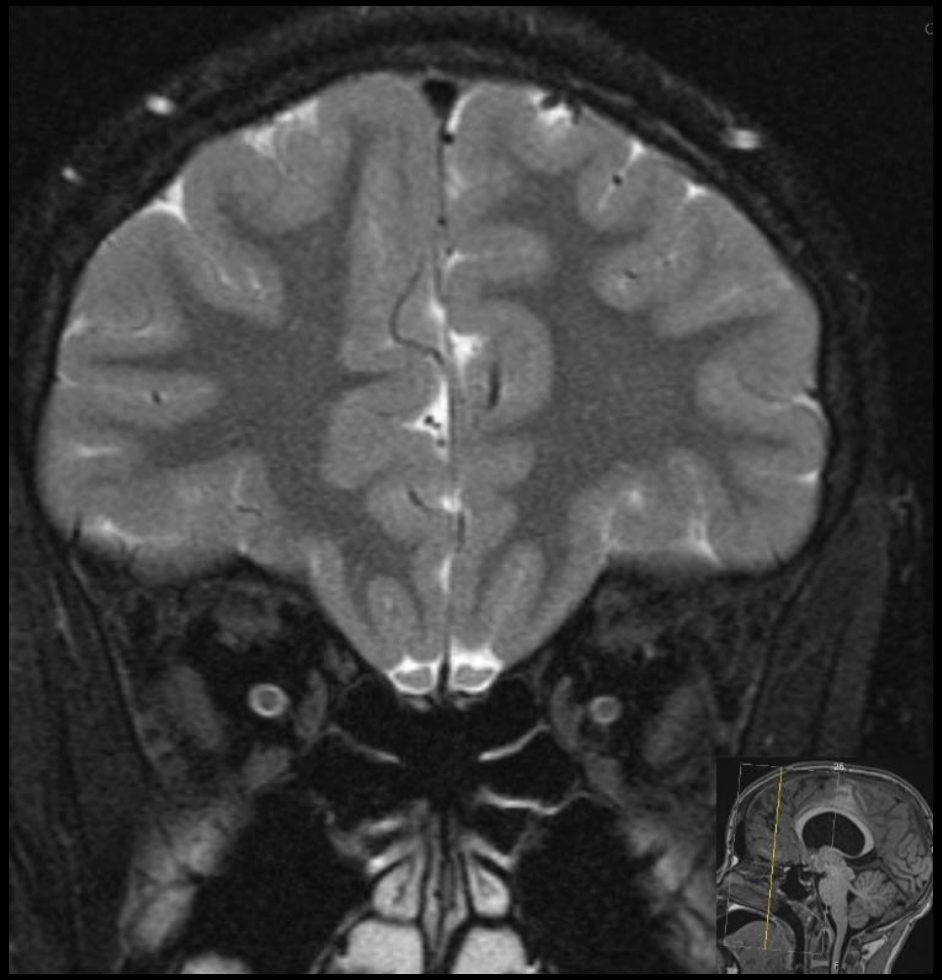
Findings (unlabeled)

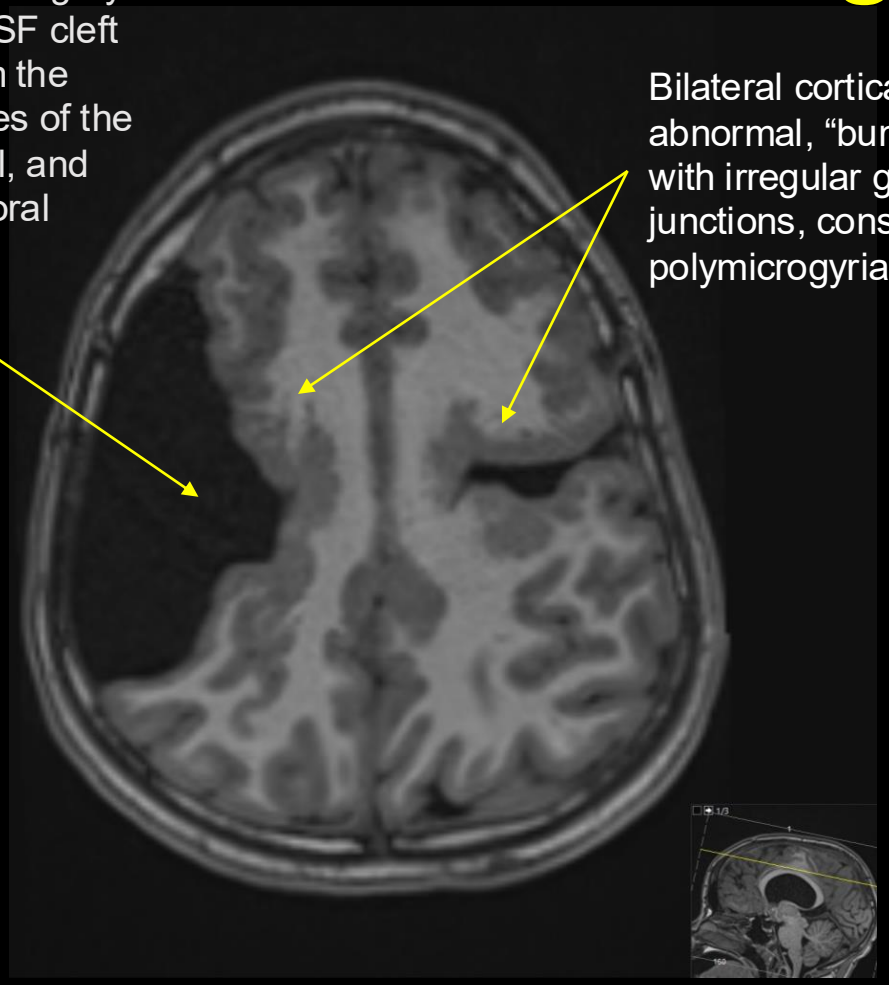


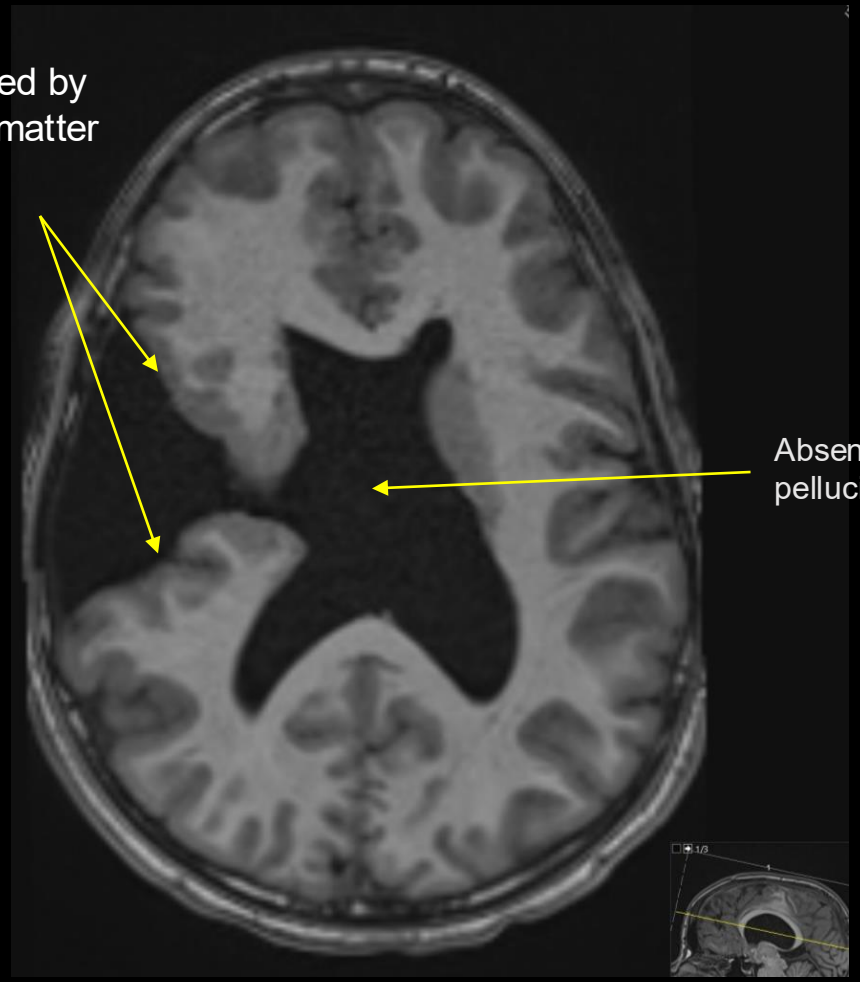
Coronal T2 FS MRI sequence

Findings (labeled)

Large right-sided gray matter lined CSF cleft extending from the cortical surfaces of the frontal, parietal, and superior temporal regions to the ventricles.

Bilateral cortical clefts lined by abnormal, “bumpy” gray matter with irregular gray-white junctions, consistent with polymicrogyria.





Absence of septum pellucidum

Axial T1 MPRAGE MRI sequence

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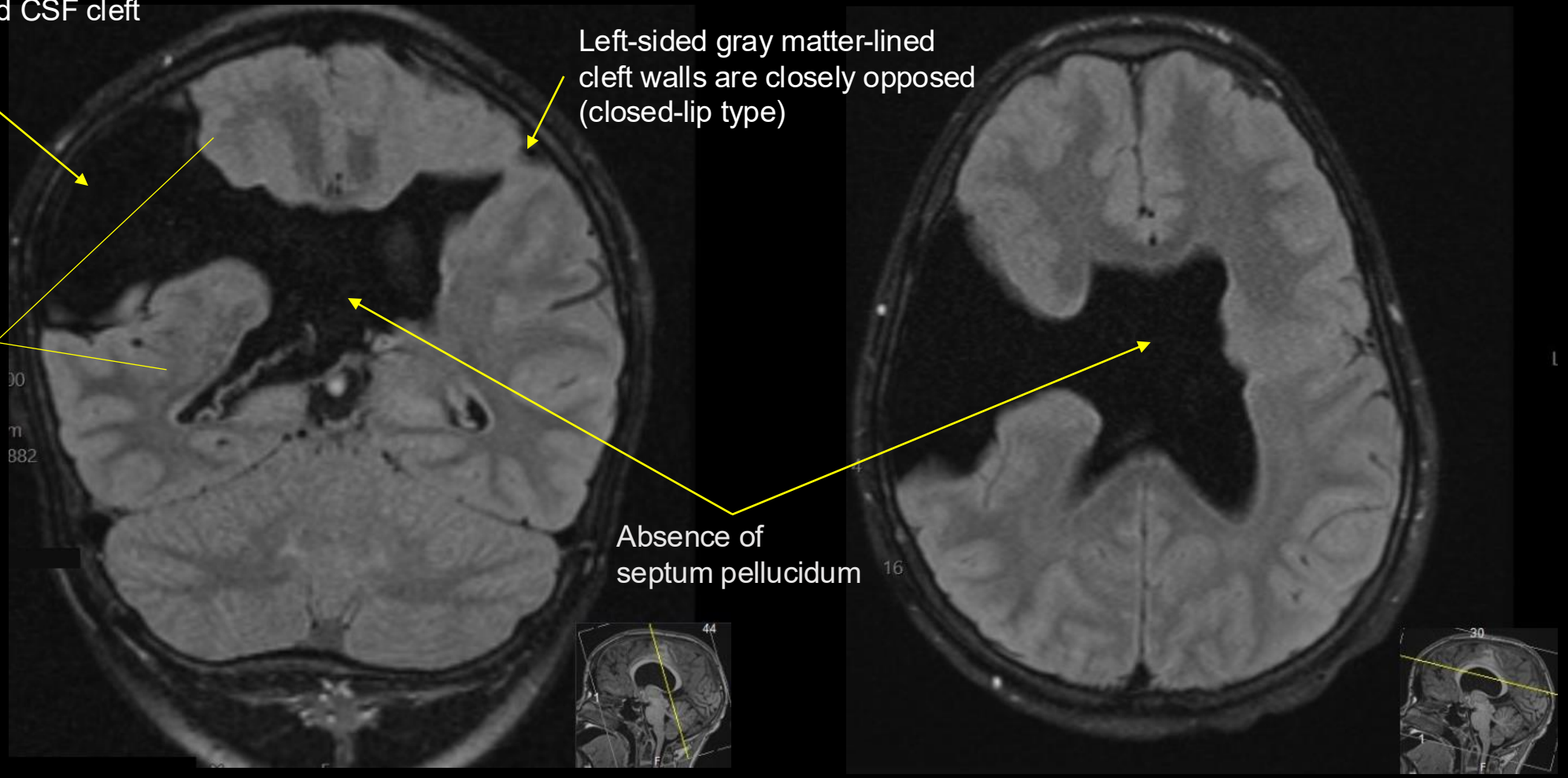
Findings (labeled)

Wide right-sided transmantle gray matter-lined CSF cleft (open-lip type)

Left-sided gray matter-lined cleft walls are closely opposed (closed-lip type)

"Note: The findings of polymicrogyria are less well demonstrated on FLAIR compared to MPRAGE (3D T1 sequence)

Absence of septum pellucidum



Left: Coronal FLAIR MRI Sequence
Right: Axial FLAIR MRI Sequence

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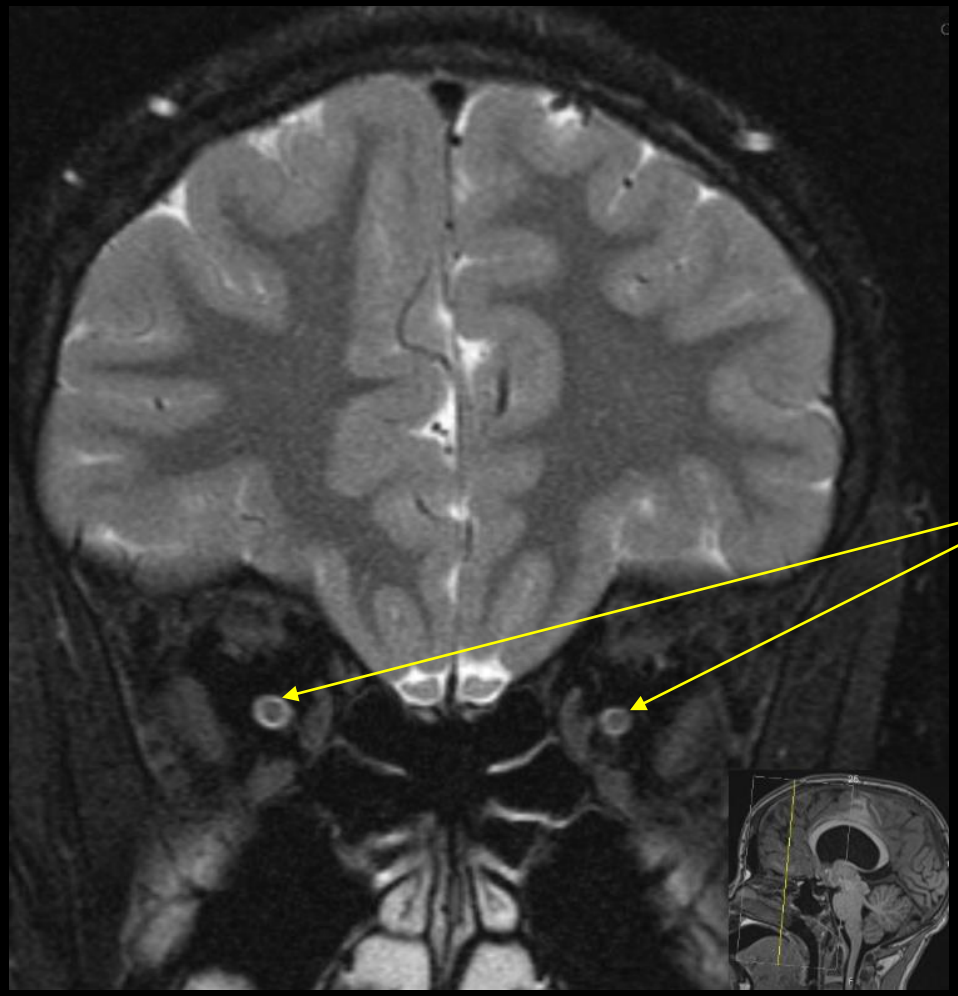
Findings (labeled)



Asymmetric optic nerves (left smaller than right), suggestive of septo-optic dysplasia in the setting of absent septum pellucidum. Seen in ~30-70% of cases of schizencephaly, with frequencies varying by study cohort. [1]

Coronal fat-saturated T2 MRI sequence

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Final Dx:

Bilateral schizencephaly, open-lip (right) and closed-lip (left)

Case Discussion: Schizencephaly

- Schizencephaly is a rare congenital cerebral malformation characterized by a cerebrospinal fluid-filled cleft extending from the pial surface to the ventricular ependyma, lined by abnormal gray matter.[1]
- Its incidence is estimated at 0.5-1.5 per 100,000 live births, with only a few familial cases reported.[2]
- First described in 1886, the etiology is still poorly understood. Most hypotheses point to anomalous neuronal migration with sparse genetic associations. [3]

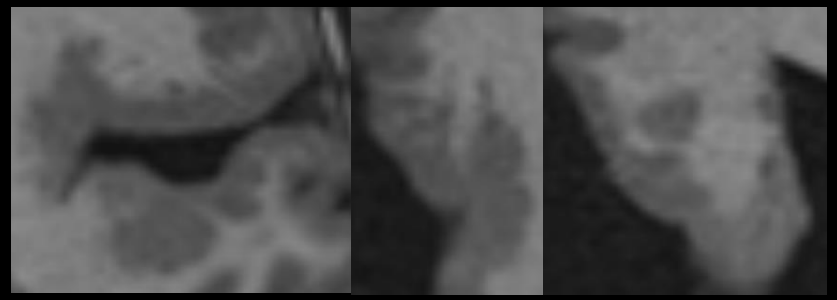
Imaging: MRI is the preferred modality, allowing differentiation of gray and white-matter lining the cleft. CT is less effective due to poorer differentiation from other fluid-associated abnormalities, such as porencephaly, where the clefts are lined by white matter. [1,4]

Differential Diagnosis[1]

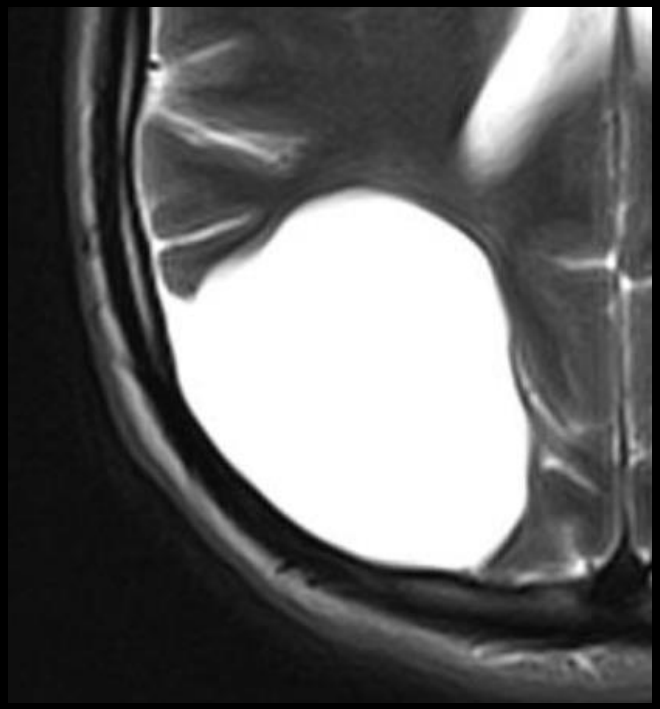
- Porencephaly
- Arachnoid Cyst
- Holoprosencephaly

*Gray matter–lined clefts extending from the ventricle to the pial surface are highly characteristic of Schizencephaly and differentiate this diagnosis from its differential considerations

Differential Diagnosis (continued)



Select magnified T1 MPRAGE images of the case presented, demonstrating dysplastic gray-matter lining the clefts consistent with schizencephaly.



Case of porencephaly demonstrating gliotic white-matter lined CSF cyst on T2 MRI.[5]

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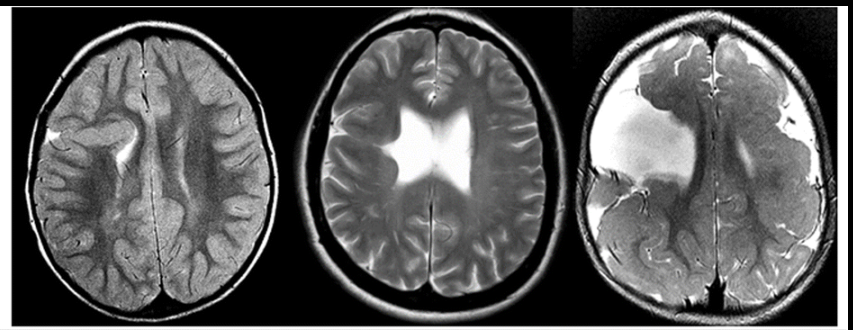
Case Discussion (continued)

- Although schizencephaly was historically described as having two types, contemporary MRI-based studies recognize three distinct classifications.[4,6]
- Type 1 - Transmantle
  - No clear CSF-filled cleft visible. Characterized by a column of abnormal gray matter stretching from the ventricle toward the cortical surface. Represents a subtle form of cortical disruption with minimal overt cleft appearance. [1,4,6]
- Type 2 - Closed-lip
  - Gray-matter-lined cleft walls are in contact or nearly touching. Cleft may contain a small amount of CSF, but the space is minimal. Often associated with milder neurological symptoms compared to open-lip forms. [1,4,6]
- Type 3 - Open-lip
  - Wide, CSF-containing cleft with separated gray-matter-lined walls. Extends continuously from the ventricular surface to the pial surface. Typically correlates with more pronounced motor deficits and epilepsy. [1,4,6]

Case Discussion (continued)



Nomenclature used in this article	Schizencephaly (type 1)	Schizencephaly (type 2)	Schizencephaly (type 3)
Nomenclature if a cleft is required for the diagnosis of schizencephaly	Trans-mantle heterotopion	Closed lip schizencephaly	Open lip schizencephaly
Nomenclature if a cleft is NOT required for the diagnosis of schizencephaly	Closed lip schizencephaly	Open lip schizencephaly	Open lip schizencephaly

Figure: MRI classification of schizencephaly showing trans-mantle, closed-lip, and open-lip types. Adapted from Griffiths PD. Neuroradiology.[6,7]

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Case Discussion (continued)

- Schizencephaly is frequently associated with additional CNS abnormalities, several of which were present in our case. In the largest systematic review to date, Braga et al. analyzed 734 patients from 156 published studies and reported: [4,7]
  - Septo-optic dysplasia: 69.1%
  - Ventricular dilatation/ventriculomegaly: 60.5%
  - Corpus callosum dysgenesis/hypoplasia: 38.2%
- Other associated malformations have been reported across multiple studies, although their frequencies vary considerably by cohort and were not consistently quantified. [4,7]
  - Absent septum pellucidum
  - Forniceal abnormalities
  - Microcephaly

Clinical Presentation

- Clinical presentation in schizencephaly is highly variable and largely determined by the size, type, and laterality of the clefts.[1,4]
- Patients may demonstrate a spectrum of neurological outcomes, from near-normal cognitive function to severe intellectual disability, depending on the anatomical extent of the malformation. [1]
- Motor deficits are among the most common neurologic findings, typically manifesting as contralateral hemiparesis in unilateral clefts and quadriparesis or more global motor dysfunction in bilateral forms. [1,4]
- Seizures are another frequent manifestation, occurring in a large proportion of patients; onset is most common in infancy or childhood but may be delayed into adolescence or adulthood, especially in those with smaller or unilateral defects.[1,4,7]

Treatment

- Management is primarily conservative and symptomatic, focusing on seizure control and physical therapy for motor deficits. [1,4,7]
- Surgical shunting may be indicated in cases of hydrocephalus or raised intracranial pressure. [1,4,7]

References:

1. Veerapaneni P, Veerapaneni KD, Yadala S. Schizencephaly. In: StatPearls. Treasure Island (FL): StatPearls Publishing; July 31, 2023.

2. Hung PC, Wang HS, Chou ML, et al. Schizencephaly in children: A single medical center retrospective study. Pediatr Neonatol. 2018;59(6):573-580. doi:10.1016/j.pedneo.2018.01.009

3. Yakovlev PI, Wadsworth RC. Schizencephalies; a study of the congenital clefts in the cerebral mantle; clefts with fused lips. J Neuropathol Exp Neurol. 1946;5:116-130. doi:10.1097/00005072-194604000-00003

4. Braga VL, da Costa MDS, Riera R, et al. Schizencephaly: A Review of 734 Patients. Pediatr Neurol. 2018;87:23-29. doi:10.1016/j.pediatrneurol.2018.08.001

5. Gaillard F. Porencephalic cyst. Radiopaedia. https://doi.org/10.53347/rID-15928. Accessed March 31, 2026.

6. Griffiths PD. Classification of the three different types of schizencephaly compared with other nomenclature systems with pictorial examples. Figure 1 in: Schizencephaly revisited. Neuroradiology. 2018;60(9):945–960. doi:10.1007/s00234-018-2056-7.

7. Griffiths PD. Schizencephaly revisited. Neuroradiology. 2018;60(9):945-960. doi:10.1007/s00234-018-2056-7

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