AMSER Case of the Month October 2020

19 mo female presenting with worsening neurological deficits and milestone regression

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Patient Presentation

- HPI: 19 month old previously healthy female presenting to the ED with regression of walking and talking for three months. Refusing to bear weight and no longer saying words. Increased irritability and sleeping. Decreased appetite. Appears pale to mother.
- PMHx: born via term spontaneous vaginal delivery
- PSHx: None
- Family Hx: 3 healthy siblings; mother with anemia requiring blood transfusions during pregnancy

Physical Exam & Pertinent Labs

- Physical Exam
  - General: mild distress
    HEENT: pale conjunctiva and lips
  - Lungs: CTAB
  - CV: regular rate and rhythm
    GI: soft, NT, liver palpable 4 cm below costal margin, palpable spleen
    Neuro: Alert, patellar clonus 4-5 beats, not cooperative with standing or sitting, normal muscle tone and bulk
- Labs: Hgb 1.3, Hct 5.0, WBC 9.8, MCV 111.1, Plt 7
- Interval history: **status epilepticus (secondary generalized seizure)**

What Imaging Should We Order?

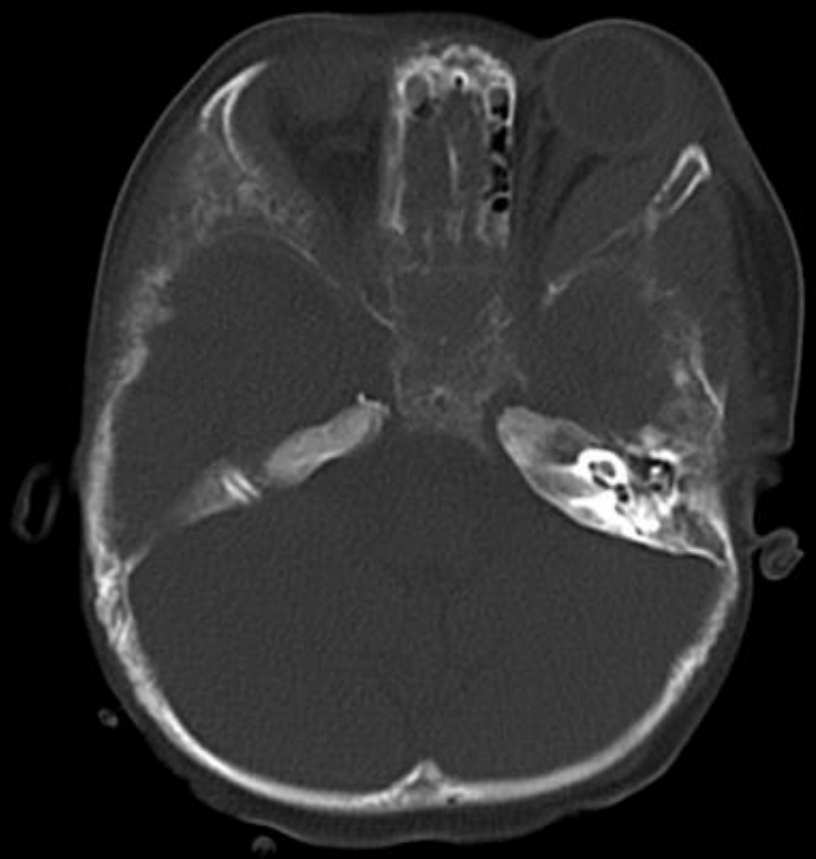
ACR Appropriateness Criteria

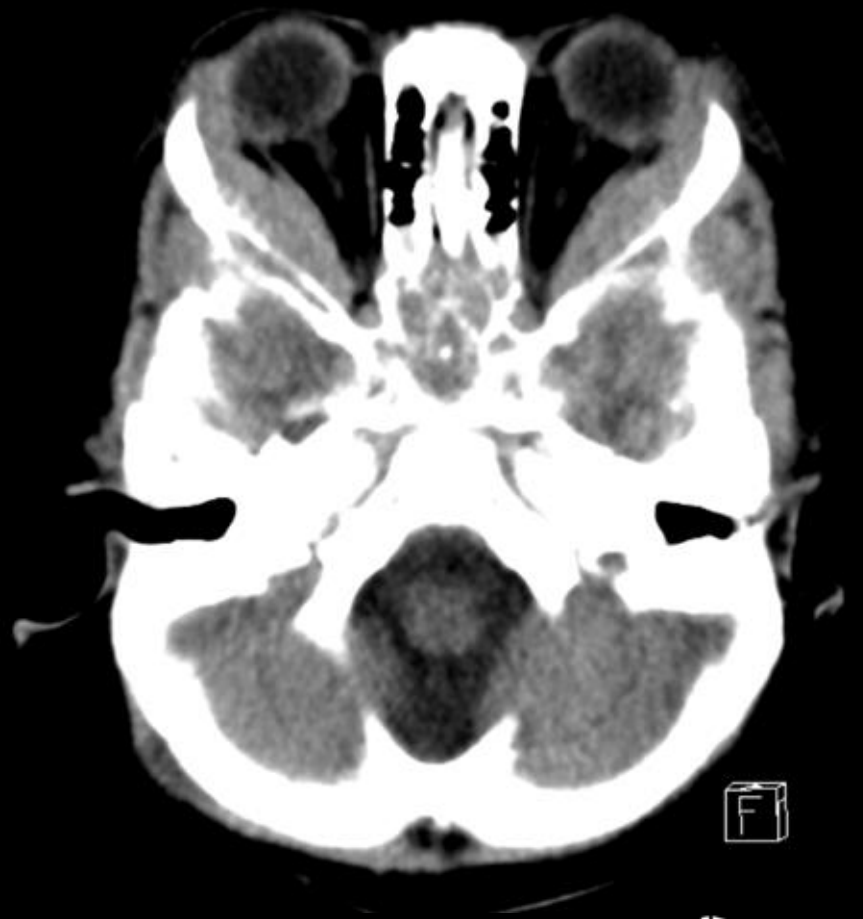
Variant 7: **Children 1 month to 18 years of age. Generalized seizure (neurologically abnormal). Initial imaging.**

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	☢☢☢☢

STAT head CT ordered by the PICU team due to status epilepticus lasting >40 mins, prompting concern for mass, bleed, or other intracranial abnormality

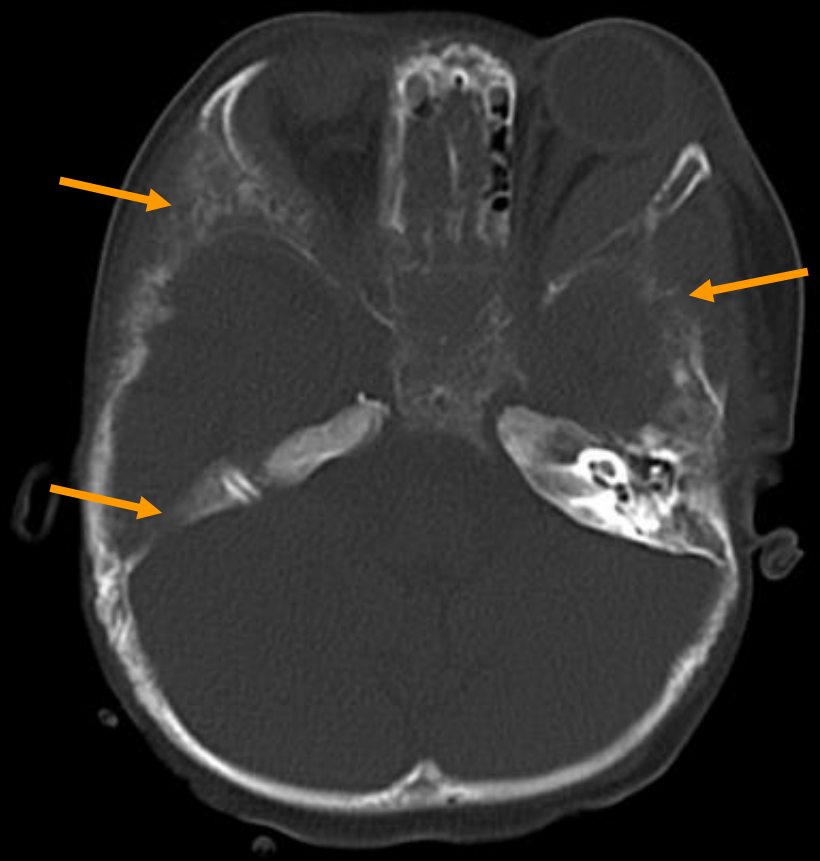
Non-Contrast Head CT





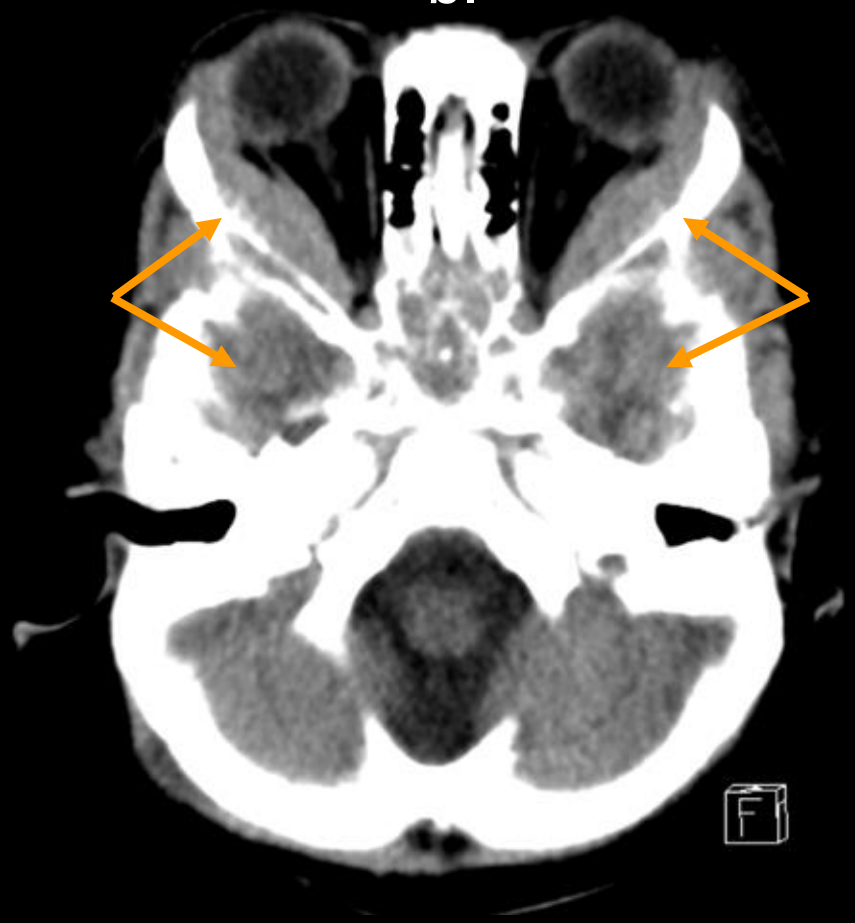
Non-Contrast Head CT

a.



Permeative-type pattern of bone loss

b.



Soft tissue mass

- a. Demineralization and permeative-type pattern of bone loss involving multiple areas throughout the skull, but most extensively involving the central skull base and greater wings of the sphenoid bone
- b. Abnormal soft tissue structure/mass centered within both greater wings of the sphenoid extending into the orbits, and both temporal and infratemporal fossas

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Further Imaging

Variant 7: **Children 1 month to 18 years of age. Generalized seizure (neurologically abnormal). Initial imaging.**

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CT head without IV contrast	May Be Appropriate	☢☢☢
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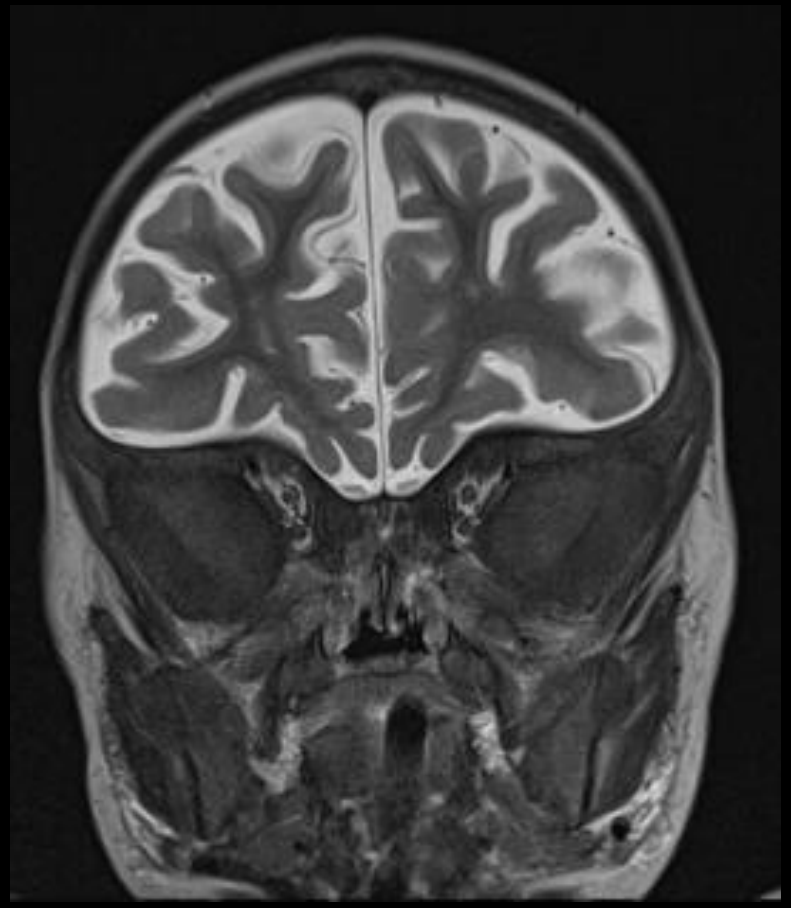
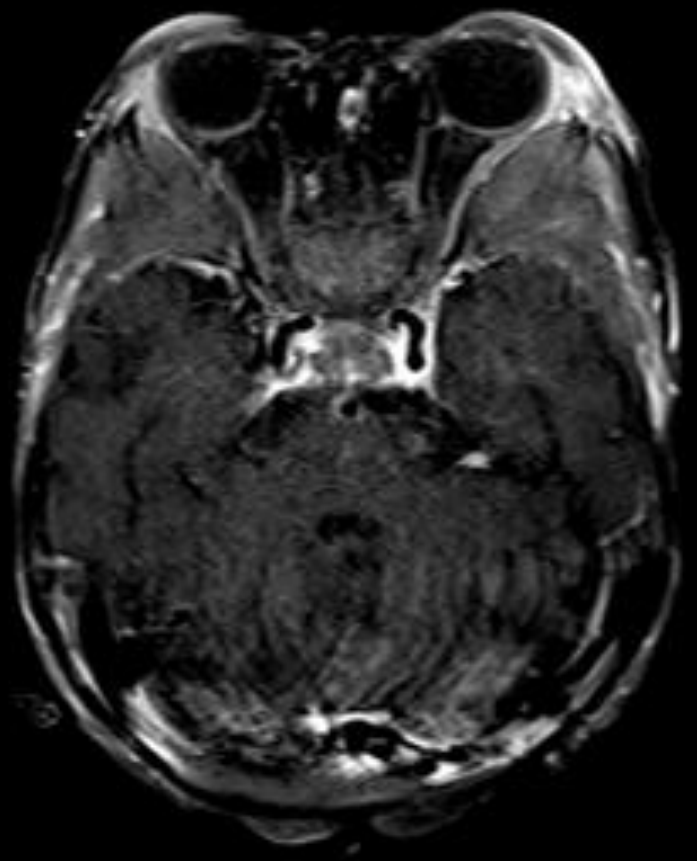
CT head with IV contrast	Usually Not Appropriate	☢☢☢
CT head without and with IV contrast	Usually Not Appropriate	☢☢☢☢
HMPAO SPECT or SPECT/CT brain	Usually Not Appropriate	☢☢☢☢
FDG-PET/CT brain	Usually Not Appropriate	☢☢☢☢

← Recommended by Neurology

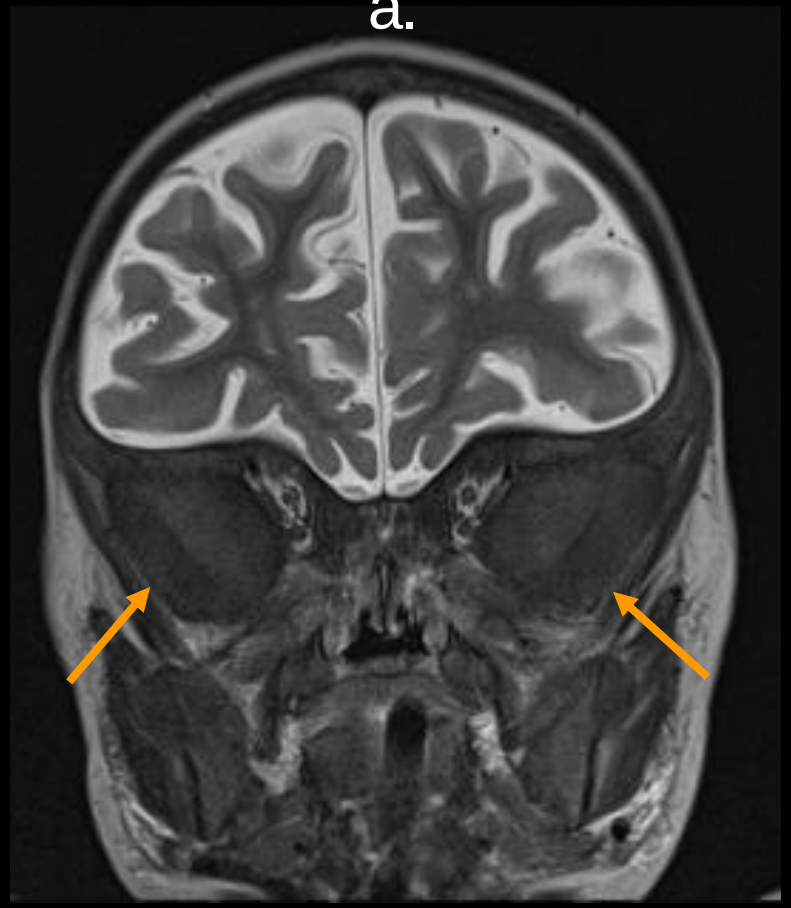
Brain MRI with/without Contrast



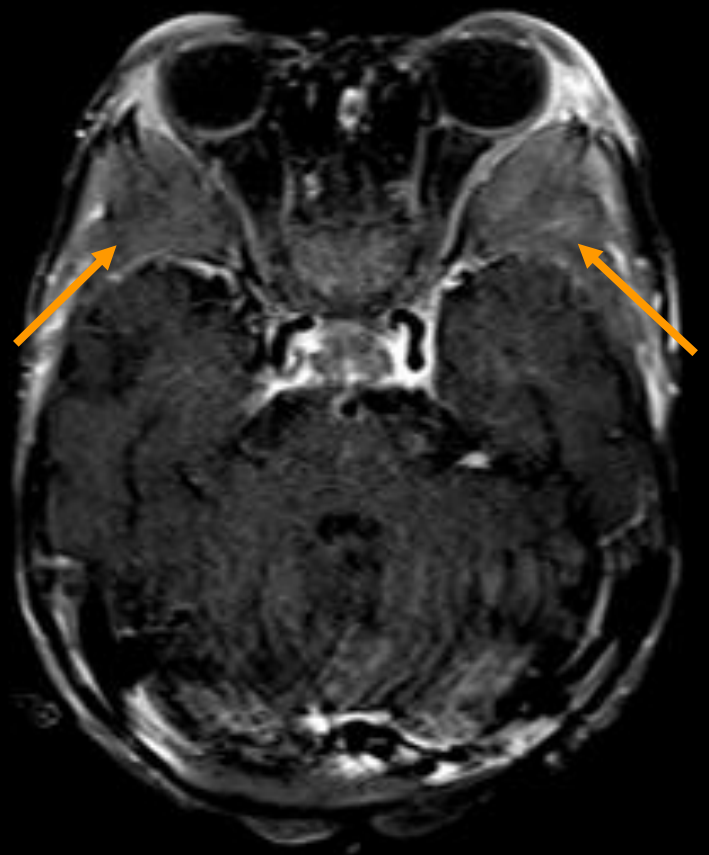


Brain MRI with/without Contrast

a.



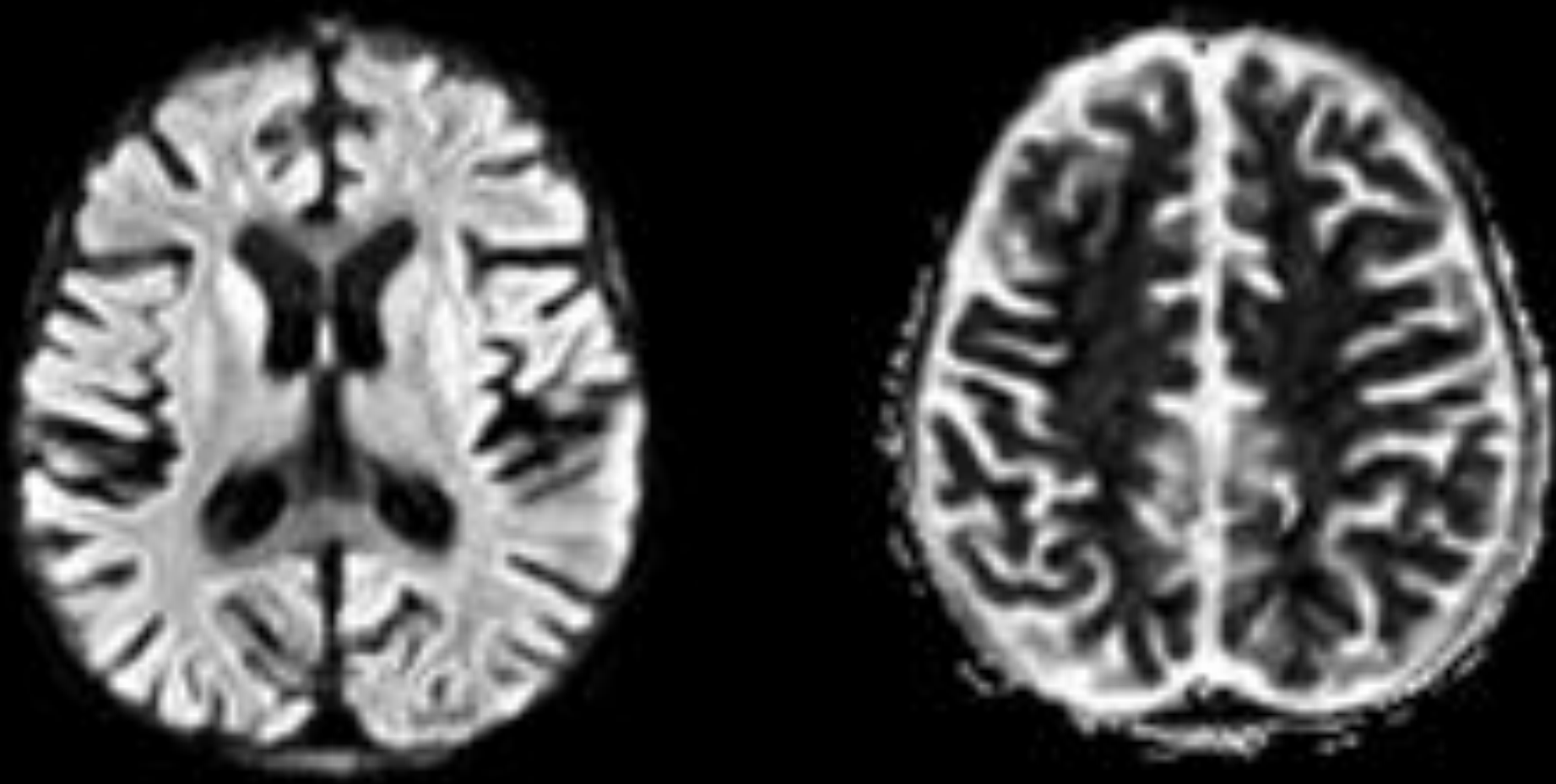
b.



- a. Generalized abnormal signal throughout the marrow space of the skull, particularly within the region of the greater wings of the sphenoid bone
- b. Abnormal enhancing soft tissue extending into the lateral aspects of the orbits, the middle cranial fossa, and the infratemporal fossa (not shown)

Soft tissue mass apparent on coronal T2 and axial T1 post views

Brain MRI with/without Contrast



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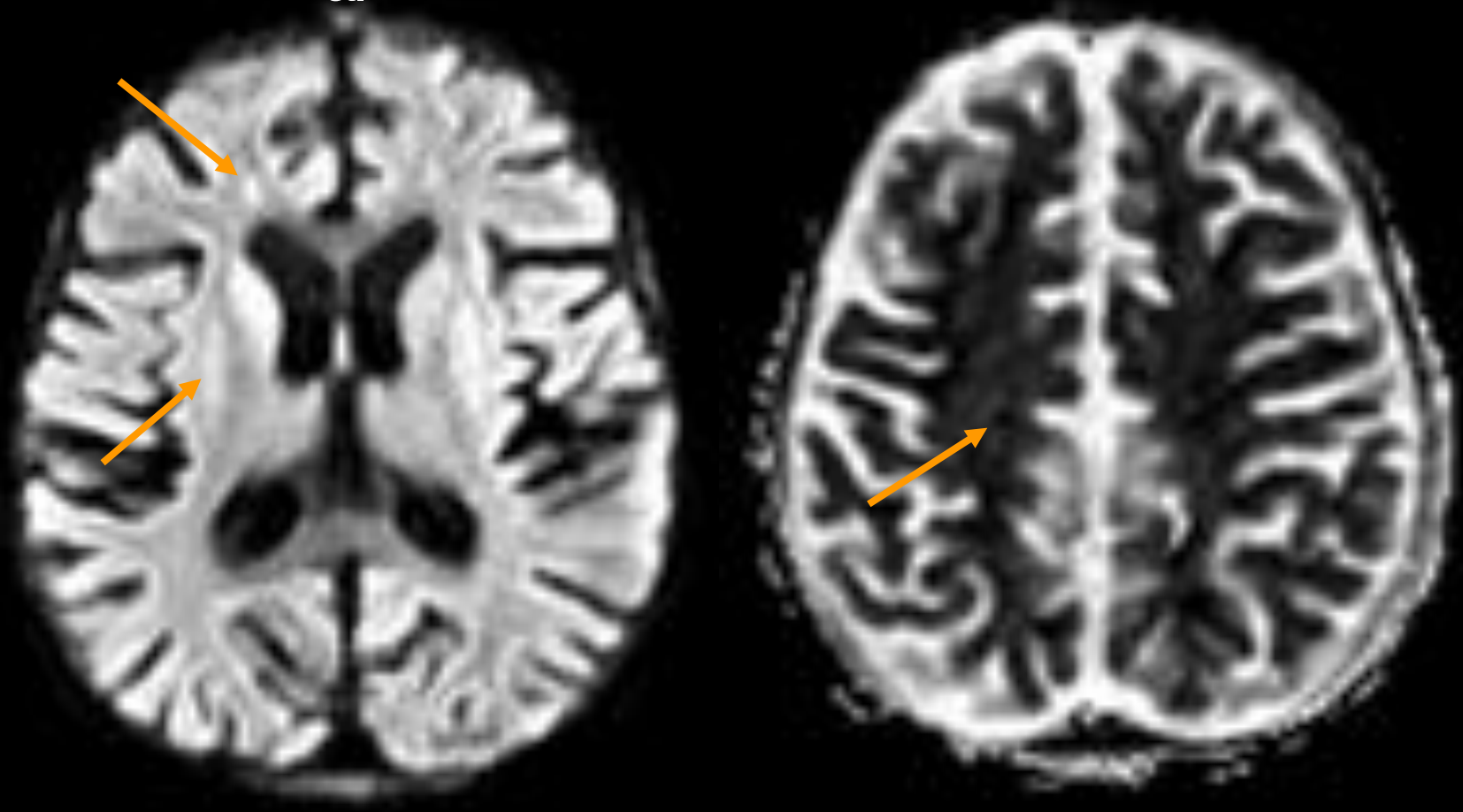
Brain MRI with/without Contrast

a.

b.



- Multiple areas of abnormal diffusion signal on DWI (a), and ADC (b, different area), that are primarily centered within the deep white matter

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Final Dx

Acute Megakaryoblastic Leukemia
complicated by watershed ischemia due to profound anemia

Acute Megakaryoblastic Leukemia (AMKL)

- Rare subset of acute myeloid leukemia that predominantly occurs in childhood; accounts for <10% of pediatric cases of AML
- May be associated with Down syndrome and t(1;22)
- Imaging may reveal evidence of diffuse marrow-replacing process with possible extension into adjacent soft tissues, as well as pachymeningeal enhancement and generalized cerebral volume loss
- Bone marrow biopsy reveals ≥20% blasts of which at least half are of megakaryocyte lineage
- Immunohistochemistry and flow cytometry aid in diagnosis

Treatment and Prognosis

- Treatment
  - Traditional chemotherapies used for other AML subtypes
  - Role of hematopoietic stem cell transplant is under investigation
- In general, AMKL is a poor individual prognostic factor for overall survival in AML
  - Exception: patients with Down syndrome have excellent prognosis (long-term survival rates >80%; complete remission rates >90%)
  - Without Down syndrome, estimated 5-year overall survival of 10%
  - t(1;22) may confer favorable prognosis

References

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