AMSER Case of the Month April 2025

5 y/o female with posterior head mass

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Patient Presentation

- HPI:
  - A 5-year-old female presents to the neurology clinic with a bulging mass on the posterior portion of her head
  - Her grandparents, who are the primary caregivers, state that she has had normal development for as long as they have known her from around 6 months old
  - They deny any headaches, seizures, or other neurological deficits
  - The mass is painless and appears to be pulsatile
  - Prenatal and previous childcare history are not well known
  - She currently attends kindergarten and does very well at school with no developmental delays reported

Pertinent Labs

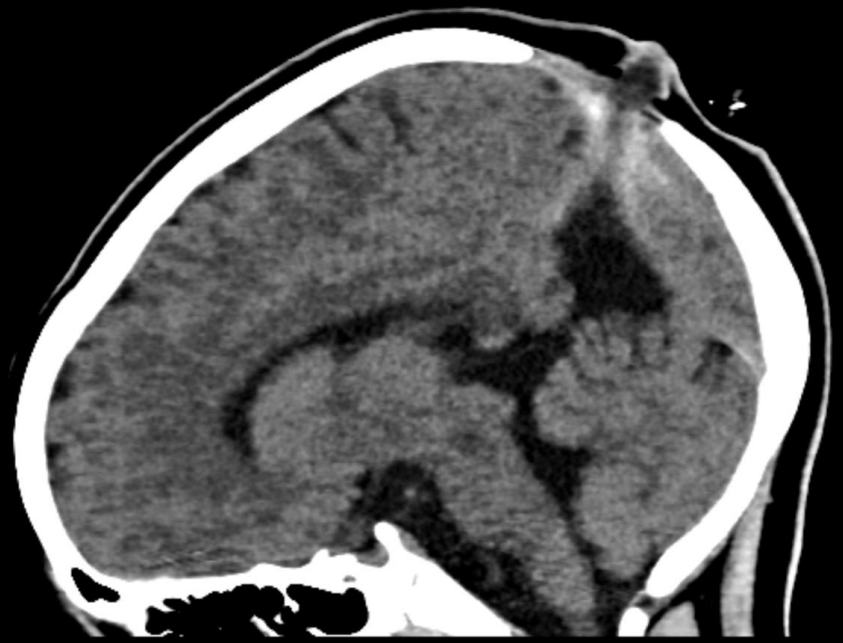
- PMHx and Surgical History: Normal developmental milestones
- Family History: Grandparents deny any history of neurological diseases
- Physical Exam:
  - Normal complete neurological exam
  - Normal muscle strength in upper and lower extremities
  - Sensory exam intact
  - Soft area over the posterior fontanelle with small skin bump
- Vitals: None
- Labs: None

What Imaging Should We Order?

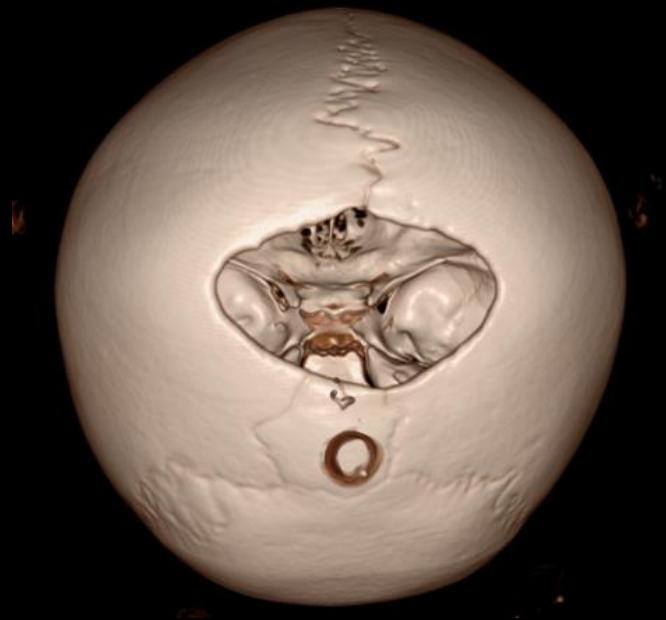
Select the applicable ACR Appropriateness Criteria

- While there is no definitive ACR criteria that is applicable to this clinical scenario, the neurologist opted to perform a CT to initially evaluate the skull abnormality
- Once the initial scans were completed and read, a follow-up MRI was performed to further characterize and define the skull abnormality

Findings (Unlabeled)

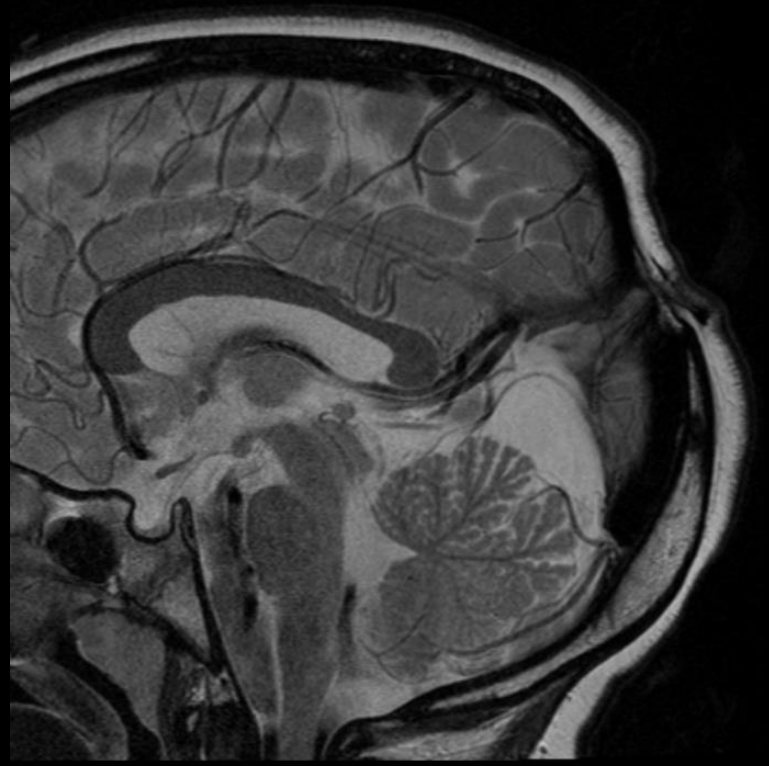


Sagittal midline CT

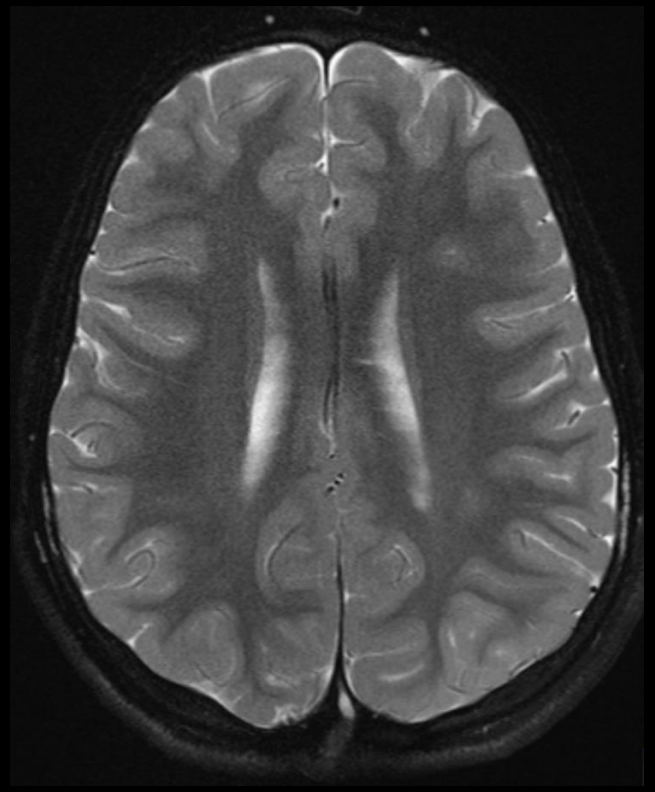


Volume rendered image of the skull (Parietal Bone)

Findings (Unlabeled)



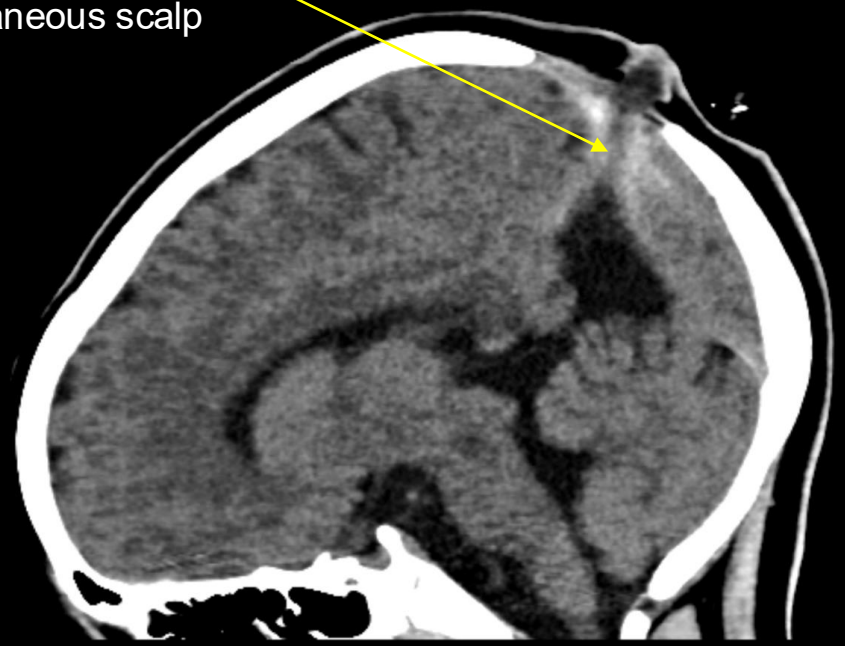
Sagittal midline T2-Weighted MRI



Axial T2-Weighted MRI

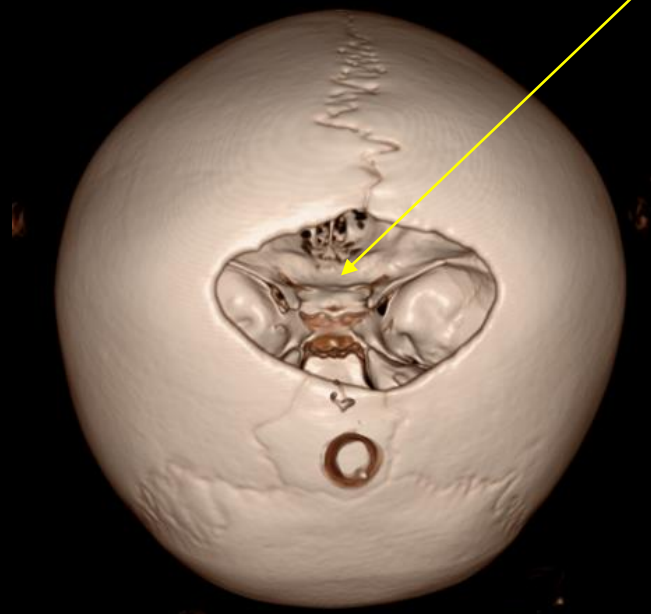
Findings (Labeled)

Cerebrospinal fluid tract extending from supracerebellar cistern through calvarial defect into subcutaneous scalp



Sagittal midline CT

Large well-defined defect within the posterior parietal bone that spans the midline



Volume rendered image of the skull (Parietal Bone)

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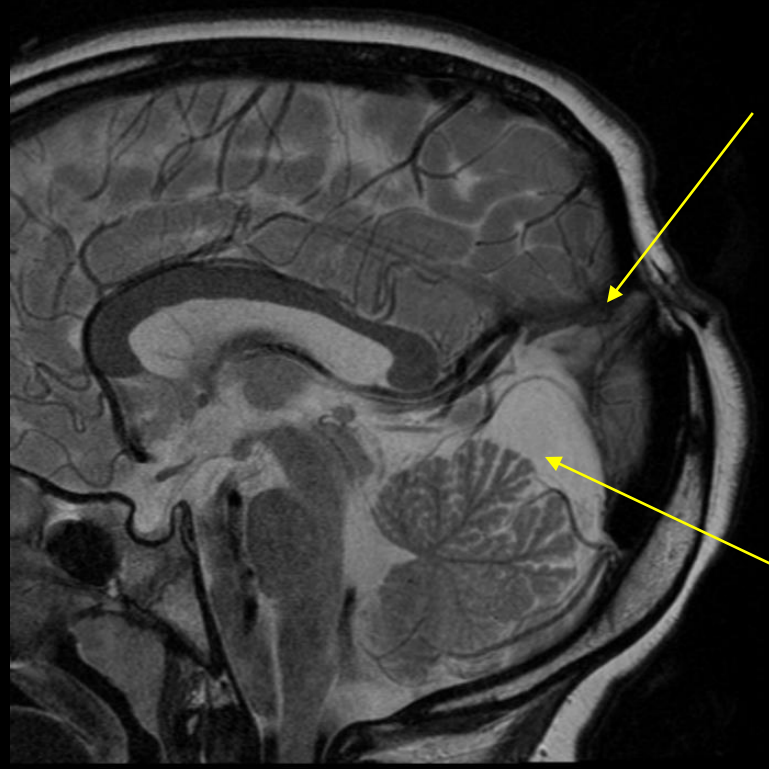
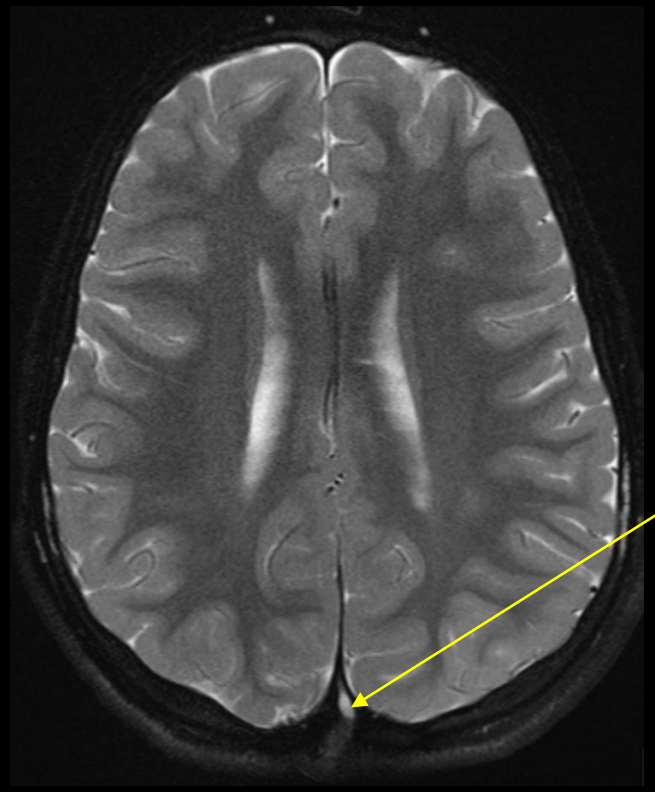


Hypointense vertical falcine vein above the cerebrospinal fluid tract

The straight sinus, usually located above the cerebellum, is absent

Sagittal midline T2-Weighted MRI



Hyperintense cerebrospinal fluid tract passing through the superior sagittal sinus

Axial T2-Weighted MRI

Final Dx:

Atretic Parietal Cephalocele

Case Discussion

- Background[1]:
  - Atretic cephaloceles are herniations of intracranial structures through a skull defect
  - It is a skin covered subgaleal lesion that contain the meninges, neural rests, and glial rests
  - Can be classified as a form of true cephalocele that has regressed with skin covering the defect[2]
  - Most commonly occur in the parietal region, followed by the occipital region[2]
- Epidemiology[3]:
  - The incidence of cephaloceles is approximately 1/3500 to 1/5000
  - Atretic cephaloceles represent 37.5% of all types of cephaloceles
  - Occur sporadically and are not linked with any genetic condition

Case Discussion

- Embryology:
  - While there is no confirmed etiology of how these cranial defects occur, there is a leading theory behind how the defect forms
  - During the initial stages of cranial venous development, the straight sinus is vertical in the ventral-dorsal plane[6]
  - When the cerebral hemispheres expand, the tentorium continues to flatten and acquire a horizontal course after the third month of gestation[7]
  - However, if a midline fibrous strand is present before the tentorium flattens to its final configuration, then it can interrupt the formation of the straight sinus and tentorium and cause fenestration of the superior sagittal sinus[7]

Case Discussion

- **Risk Factors:**
  - While not a definitive list specific to atretic cephaloceles, the same risk factors and preventative measures for neural tube defects apply
  - Low folate (Vitamin B9) during the first trimester of pregnancy[3]
  - Untreated pre-existing medical conditions such as diabetes[4]
  - Teratogenic medications such as antiseizure medications[4]
  - Chronically overheating such as sauna or hot tub use or frequent fevers[4]
- **Clinical Presentation[2]:**
  - Reported cases of atretic cephaloceles can appear completely normal, achieve age-appropriate developmental milestones, or develop severe developmental delays if other intracranial anomalies are present
  - The lesions themselves are typically slow growing with a hairless skin patch overlying the lesion
  - Can be pulsatile and soft upon palpation

Case Discussion

- Treatment:
  - Most cases are treatable with simple neurosurgery intervention with good outcomes
  - The cyst is excised surgically and then the tract is oversewn over the dura to facilitate proper healing[2]
  - Post surgery, most cases show normal development with no special management needed with regular follow up with pediatrics[2]
- Radiologic Findings:
  - On CT, atretic cephaloceles will show narrowing of the bone defect from inside out compared to other possible diagnoses such as dermoid cysts where the bone edges narrow from the outside in[9]
  - On MRI
    - A soft tissue mass will be seen with a sharply demarcated calvarial defect[8]
    - CSF tract and a vertical falcine vein are clear remnants of the formation process[8]
    - A fibrous stalk that connects the cephalocele can be seen with focal fenestration of the superior sagittal sinus[8]
    - The superior cerebellar cistern and suprapineal recess can be enlarged[8]
    - The posterior tentorium will peak superiorly[8]

References:

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