

AMSER Case of the Month

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85 Year old female with altered mental status and facial cutaneous hemangiomas

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Patient Presentation

- **HPI:** 85-year-old female presents to the ED after an acute fall with altered mental status and noted to have facial cutaneous hemangiomas.
- **Pertinent PMH:** Multiple complications including advanced glaucoma bilaterally with right vision loss and history of seizure disorder
- **Medications:** Phenytoin 100 mg BID, Timolol 0.5 % ophthalmic solution

Pertinent Physical Exam and Labs

- **Physical Exam:**
 - Skin: Large facial hemangiomas covering the entire face; Has scattered neurofibromas.
 - Neuro: Alert and Oriented x1 to self
- **Labs:** Non-contributory

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

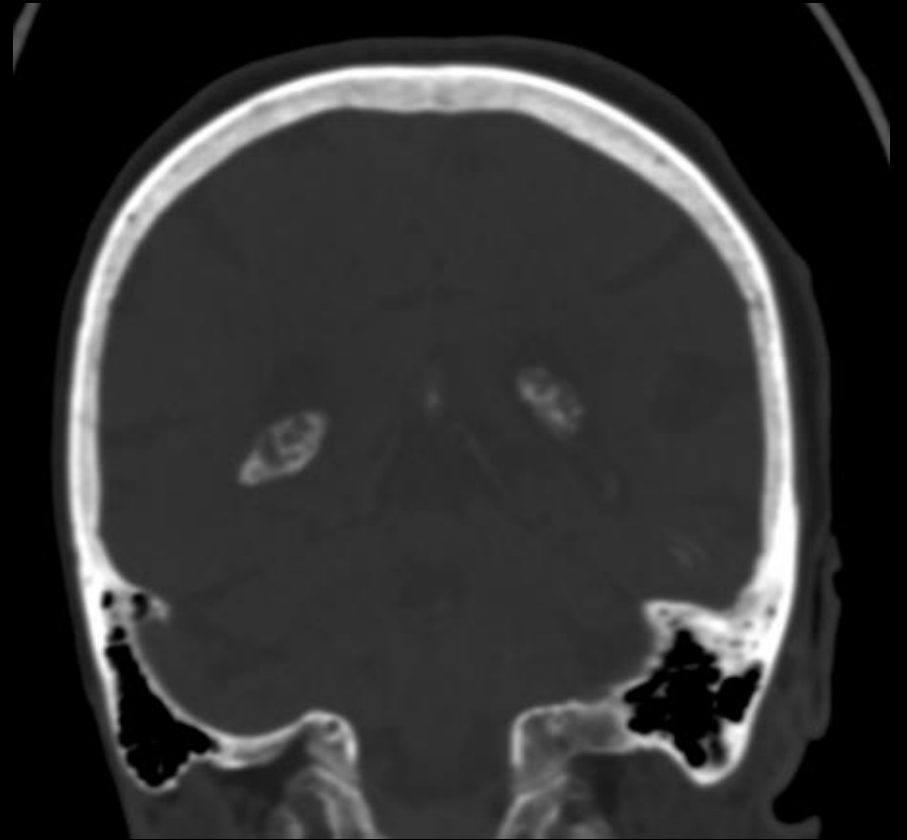
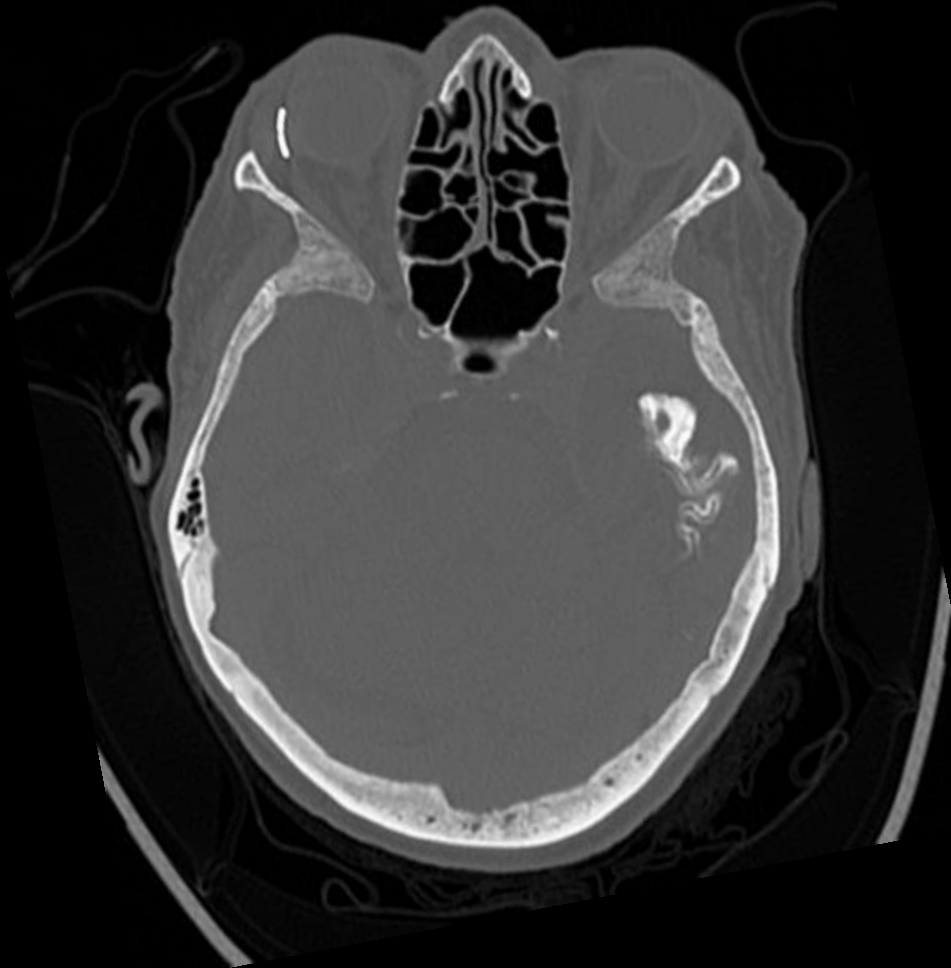
Variant 1:

Acute mental status change. Increased risk for intracranial bleeding (ie, anticoagulant use, coagulopathy), hypertensive emergency, or clinical suspicion for intracranial infection, mass, or elevated intracranial pressure. Initial imaging.

Procedure	Appropriateness Category	Relative Radiation Level
CT head without IV contrast	Usually Appropriate	☼ ☼ ☼
MRI head without IV contrast	Usually Appropriate	0
MRI head without and with IV contrast	May Be Appropriate	0
CT head without and with IV contrast	May Be Appropriate	☼ ☼ ☼
CT head with IV contrast	Usually Not Appropriate	☼ ☼ ☼

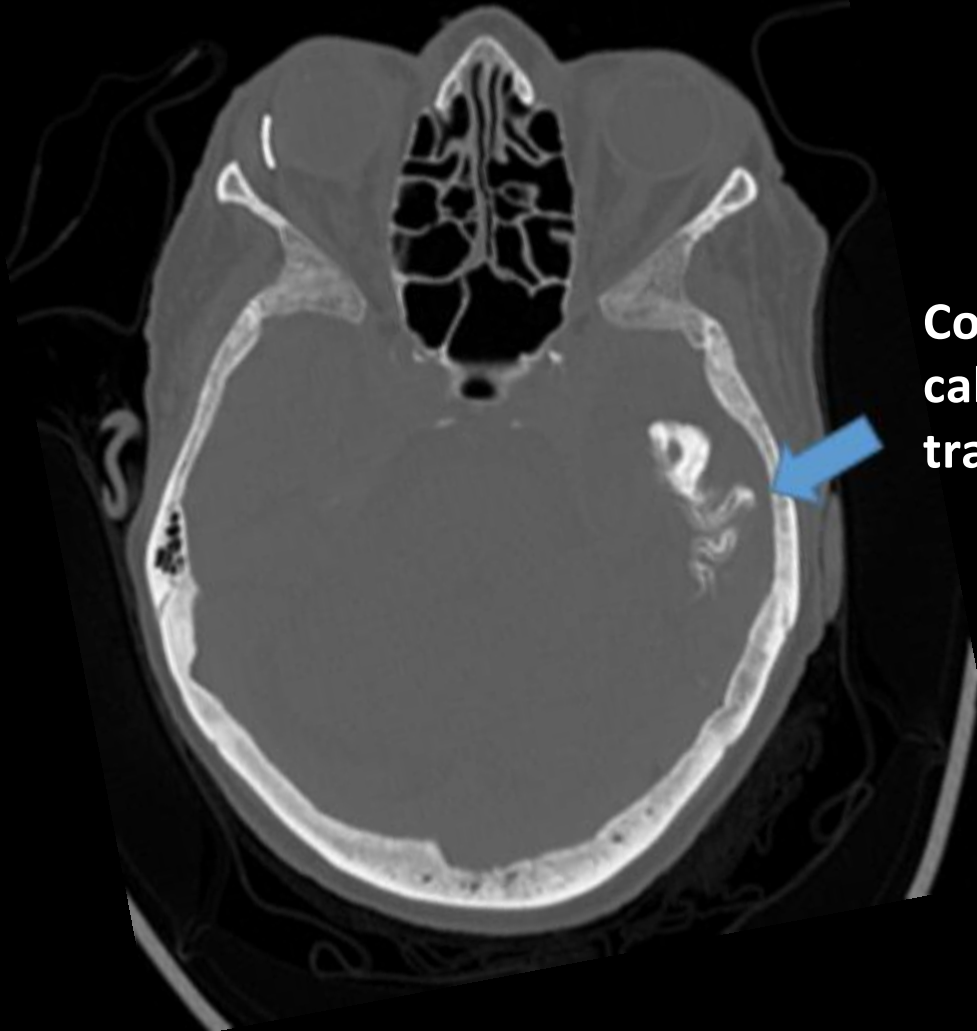
This imaging modality was ordered by the ER physician

Findings CT Head without Contrast (unlabeled)

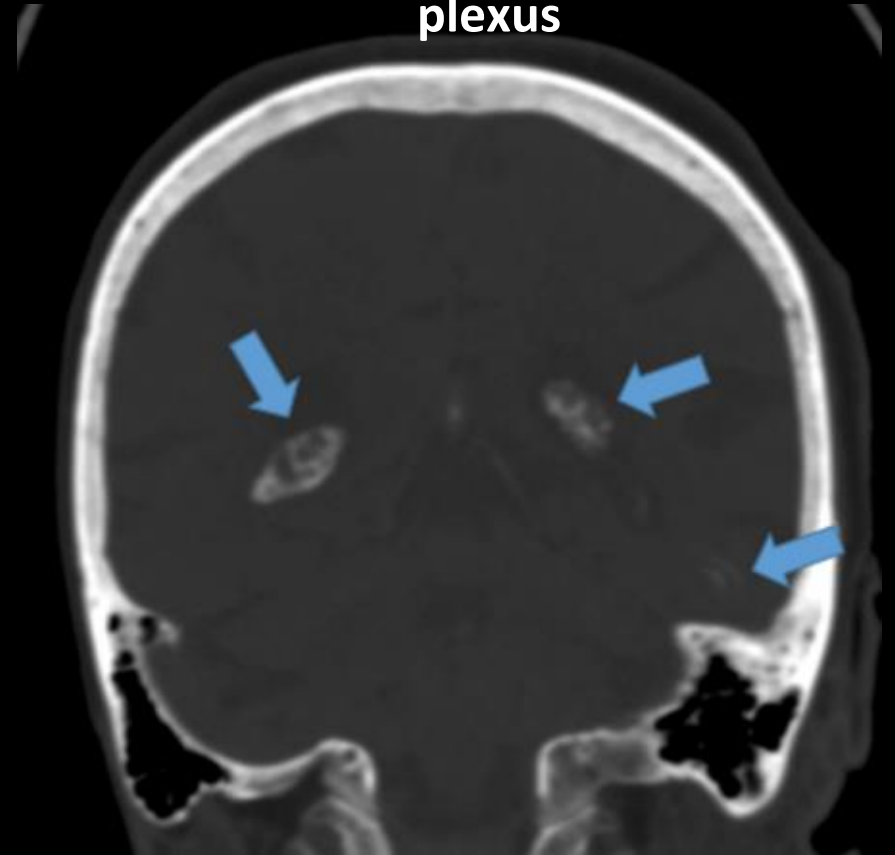


Findings: (labeled)

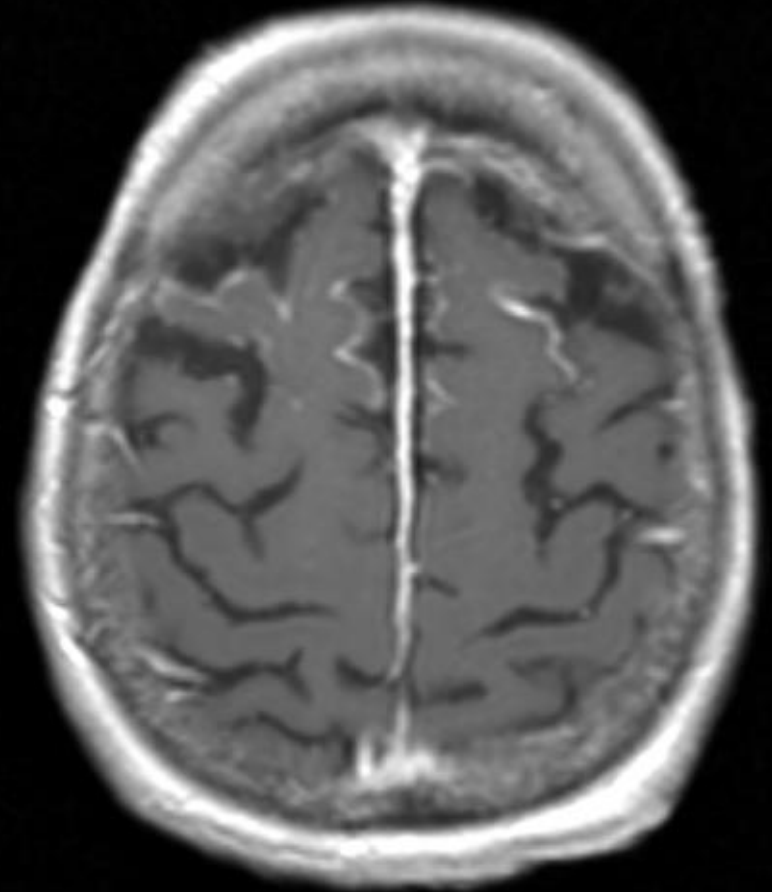
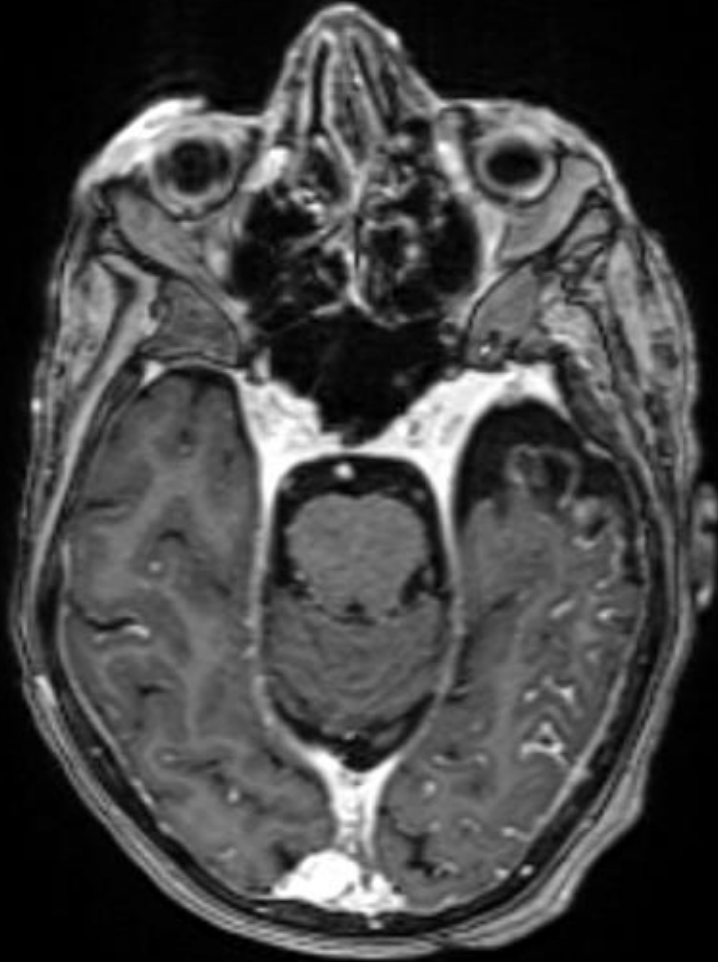
Cortical/subcortical calcifications with tram track sign



Cortical calcification and bilateral enlargement and calcification of the choroid plexus

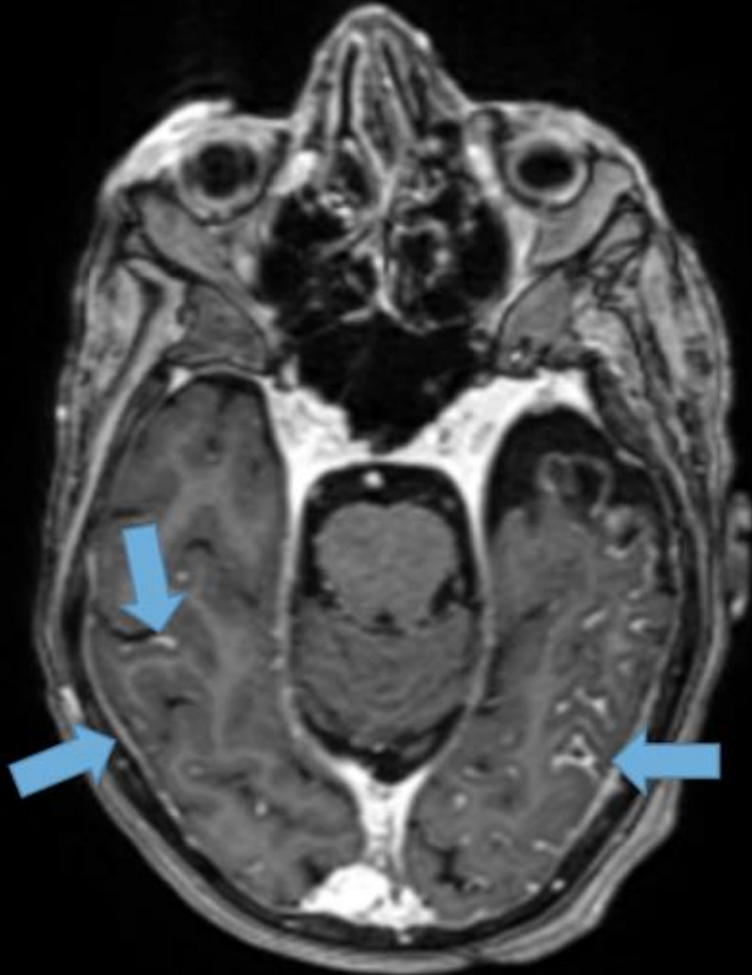


MRI T1 with Contrast Findings: (Unlabeled)

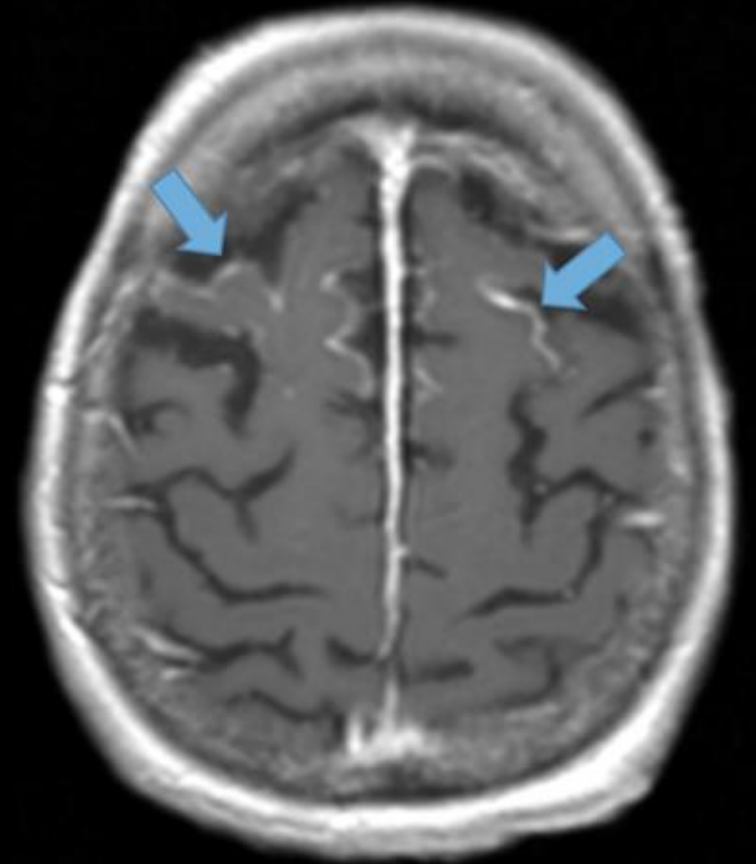


Findings: (labeled)

T1 post contrast image showing leptomeningeal enhancement



T1 post contrast image showing leptomeningeal enhancement



Final Dx:

Sturge Weber

Case Discussion

- Sturge Weber Syndrome is a neurocutaneous syndrome characterized by facial capillary malformation (port wine stain) usually seen in the distribution of the trigeminal nerve and clusters of abnormal blood vessels known as leptomeningeal angiomas
 - Caused by a mutation in GNAQ gene; the gene mutation is not inherited, but occurs by chance in cells of the developing embryo
- Clinical features include:
 - Facial capillary malformation
 - Leptomeningeal angioma involving the brain and eye
 - Seizure disorder
 - Glaucoma
 - Hemiparesis and stroke-like events

Diagnosis and Management

- **Diagnosis:**
 - The preferred neuroimaging technique for the diagnosis is brain MRI with gadolinium, which demonstrates the presence of leptomeningeal capillary venous malformation
 - CT Head non contrast can identify brain calcification and provide some anatomic information
- **Management:**
 - No specific management treatment exists for SWS
 - Treatment revolves primarily around seizure control, with surgical resection only indicated rarely in refractory cases--hemispherectomy or focal resection of the seizure focus. Patients with bilateral involvement are typically not good candidates for surgery
 - Ophthalmological examination is also essential to identify and treat ocular involvement
 - Low-dose aspirin has also been shown to be effective in decreasing the frequency of seizures and stroke-like episodes

References:

1. Comi AM. Sturge-Weber syndrome. *Handb Clin Neurol*. 2015;132:157-68.
2. Hassanpour K, Nourinia R, Gerami E, Mahmoudi G, Esfandiari H. Ocular Manifestations of the Sturge-Weber Syndrome. *J Ophthalmic Vis Res*. 2021 Jul 29;16(3):415-431.
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