

AMSER Case of the Month: November 2025

36-year-old female with acute onset of left face/arm
weakness & sensory changes

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Patient Presentation

The patient is 36 y.o. female who presented to ED in 2022 with acute onset of left-sided facial weakness/droop and tingling, left arm weakness and tingling, and a sensation of being off-balance.

At initial evaluation in ED, she reported that her symptoms had drastically improved

No significant findings on physical exam other than mild left-sided droop of the face and flattened nasolabial fold (NIHSS=1)

Past Medical History:

Patient has a history of Headaches

Examination;

Blood pressure 178/88. Patient's vital signs, mental status, neurological exam otherwise within normal limits.

Family History of Hypercoagulable disease present.

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

Variant 2: Adult. Focal neurologic deficit. Clinically suspected acute ischemic stroke. Initial imaging.

Procedure	Appropriateness Category	Relative Radiation Level
MRI head without IV contrast	Usually Appropriate	○
CT head without IV contrast	Usually Appropriate	☼☼☼
CTA head with IV contrast	Usually Appropriate	☼☼☼
CTA neck with IV contrast	Usually Appropriate	☼☼☼
US duplex Doppler carotid artery	May Be Appropriate	○
MRA head without IV contrast	May Be Appropriate	○
MRA neck without and with IV contrast	May Be Appropriate	○
MRA neck without IV contrast	May Be Appropriate	○
MRI head perfusion with IV contrast	May Be Appropriate	○
CT head perfusion with IV contrast	May Be Appropriate	☼☼☼
US duplex Doppler transcranial	Usually Not Appropriate	○
Arteriography cervicocerebral	Usually Not Appropriate	☼☼☼
MRA head without and with IV contrast	Usually Not Appropriate	○
MRI head without and with IV contrast	Usually Not Appropriate	○
MRV head without and with IV contrast	Usually Not Appropriate	○
MRV head without IV contrast	Usually Not Appropriate	○
CT head with IV contrast	Usually Not Appropriate	☼☼☼
CT head without and with IV contrast	Usually Not Appropriate	☼☼☼
CTV head with IV contrast	Usually Not Appropriate	☼☼☼

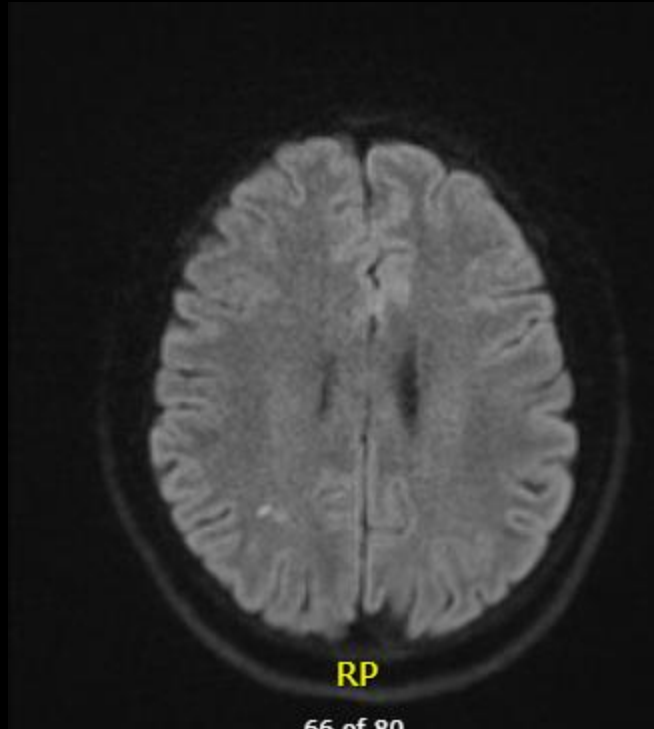
These imaging modalities were ordered by the ER physician



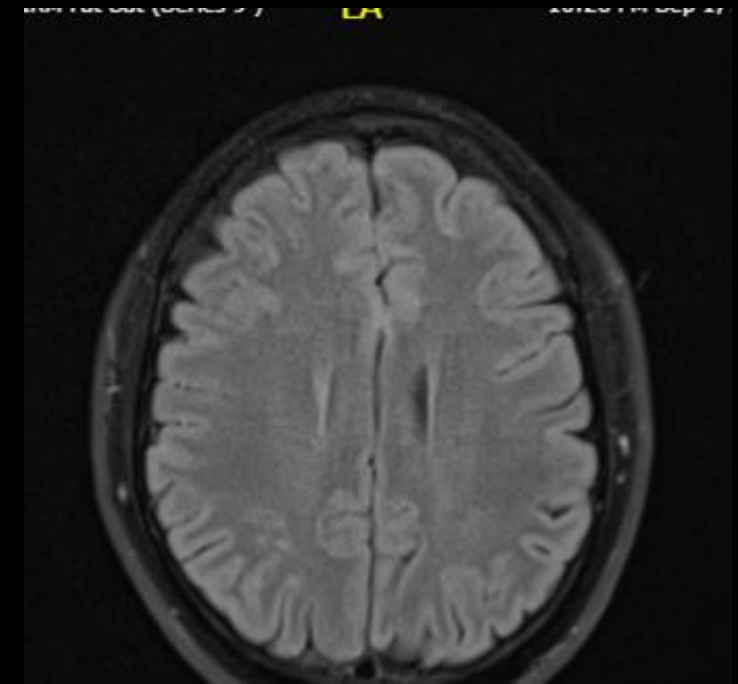
Findings (unlabeled)



CT Head WO IV Contrast



DWI

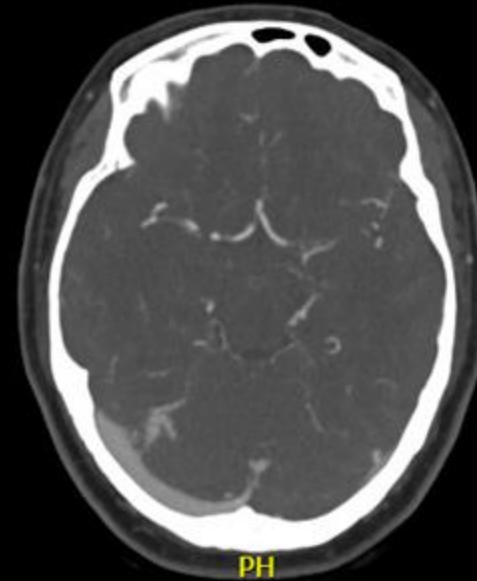
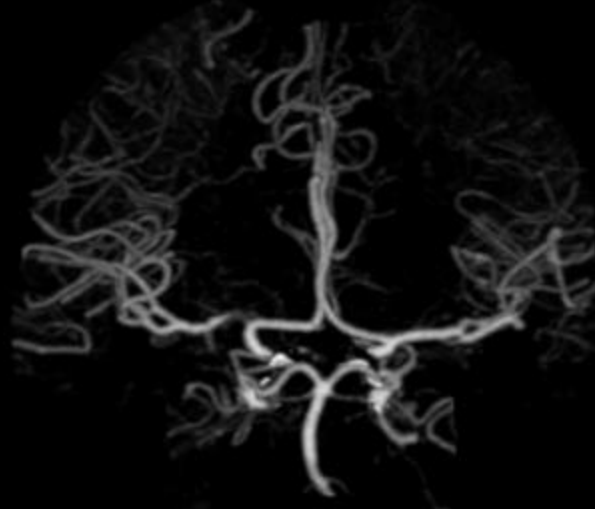
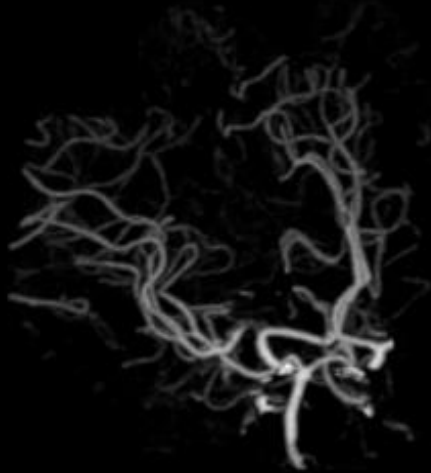


Axial FLAIR

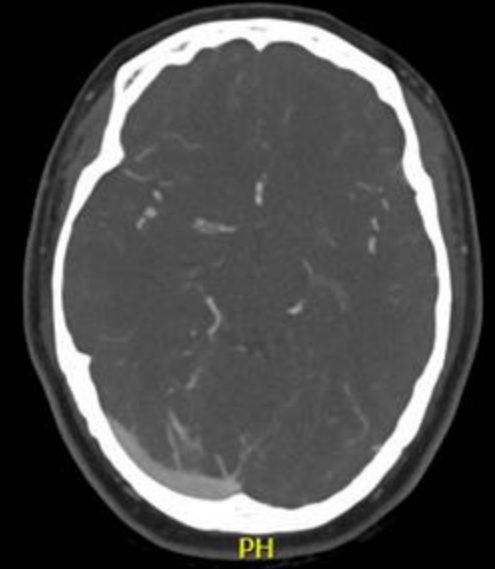
MRI Brain WO Contrast

Findings (unlabeled)

Right Hemisphere



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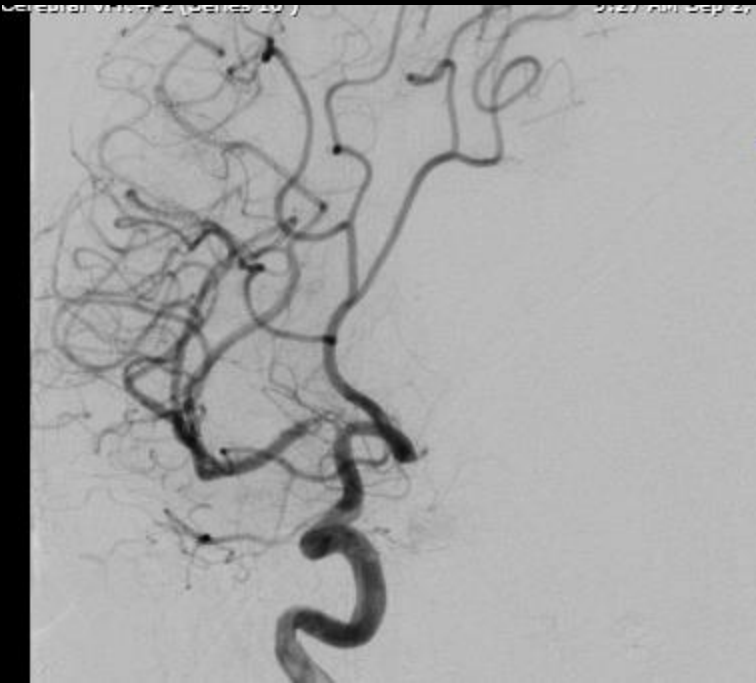


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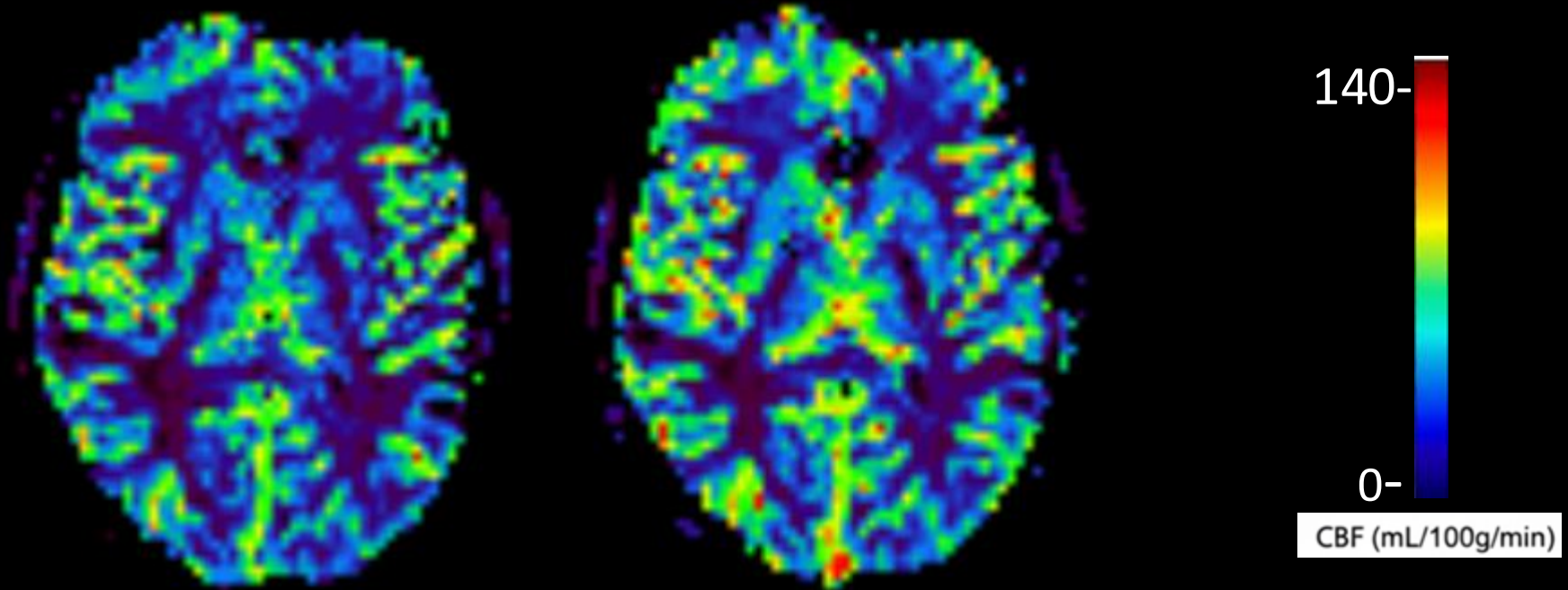
CT Angio Head w IV Contrast

Findings (unlabeled)



Diagnostic Angiogram

Findings: (unlabeled)



Cerebral Perfusion MRI Following Acetazolamide Challenge

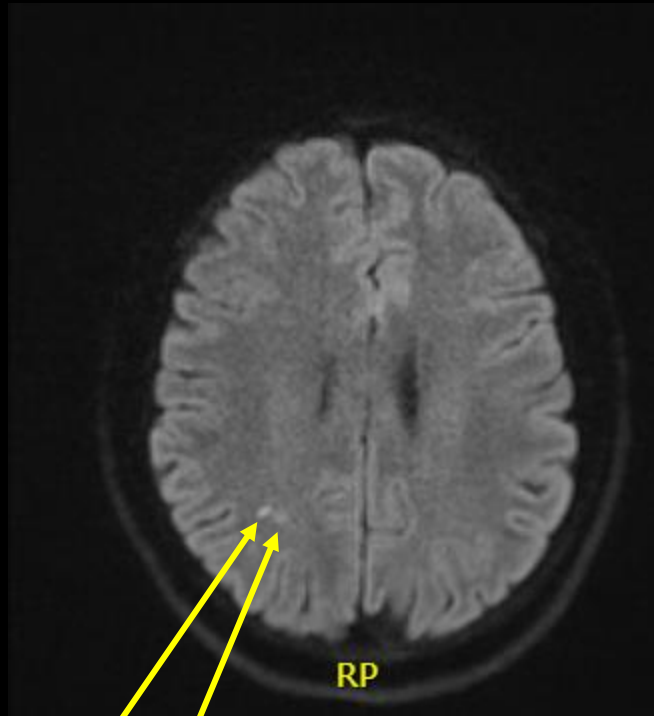
Left: Pre-challenge, Right: Post-challenge MR perfusion Cerebral blood flow (CBF) maps.

Findings (labeled)

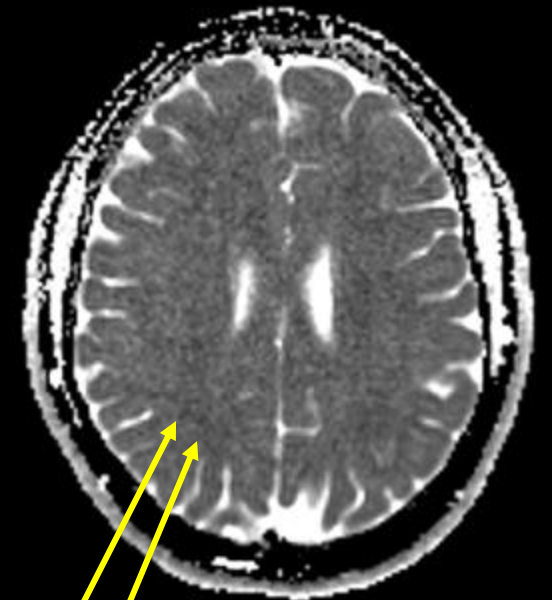


CT Head WO IV Contrast

Normal



DWI

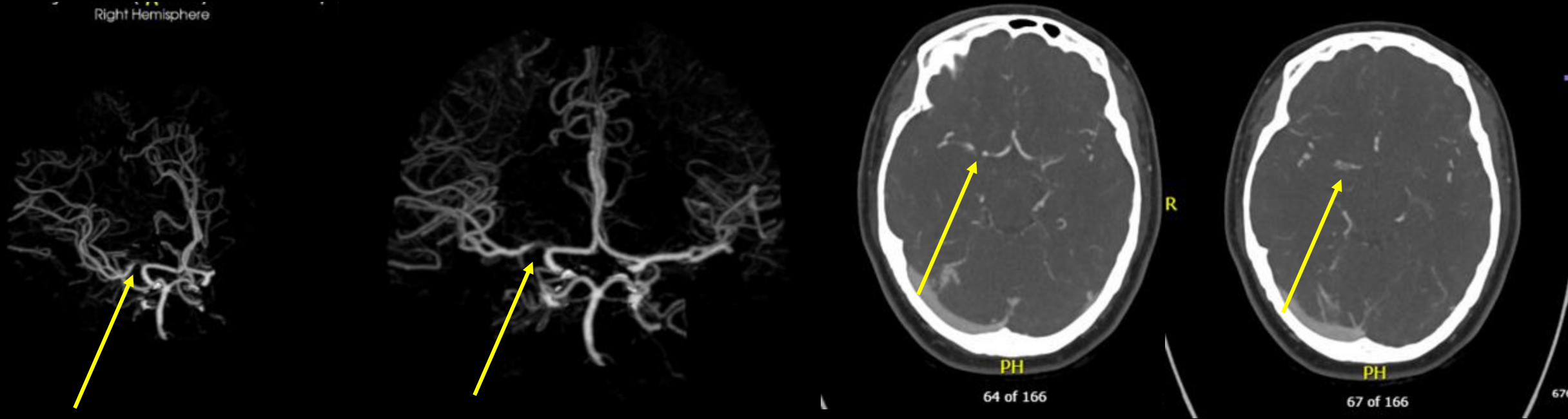


ADC

MRI Brain WO Contrast

2 tiny foci of mildly restricted diffusion new from 3 months prior, within the right parietal white matter consistent with subacute infarction.

Findings: (labeled)



CT Angio Head w IV Contrast

Focal high-grade stenosis with a network of adjacent collateral vessels at the proximal right M1 segment versus, less likely, subocclusive filling defect with surrounding marginal flow. The 4th image shows a tuft of smoke appearance

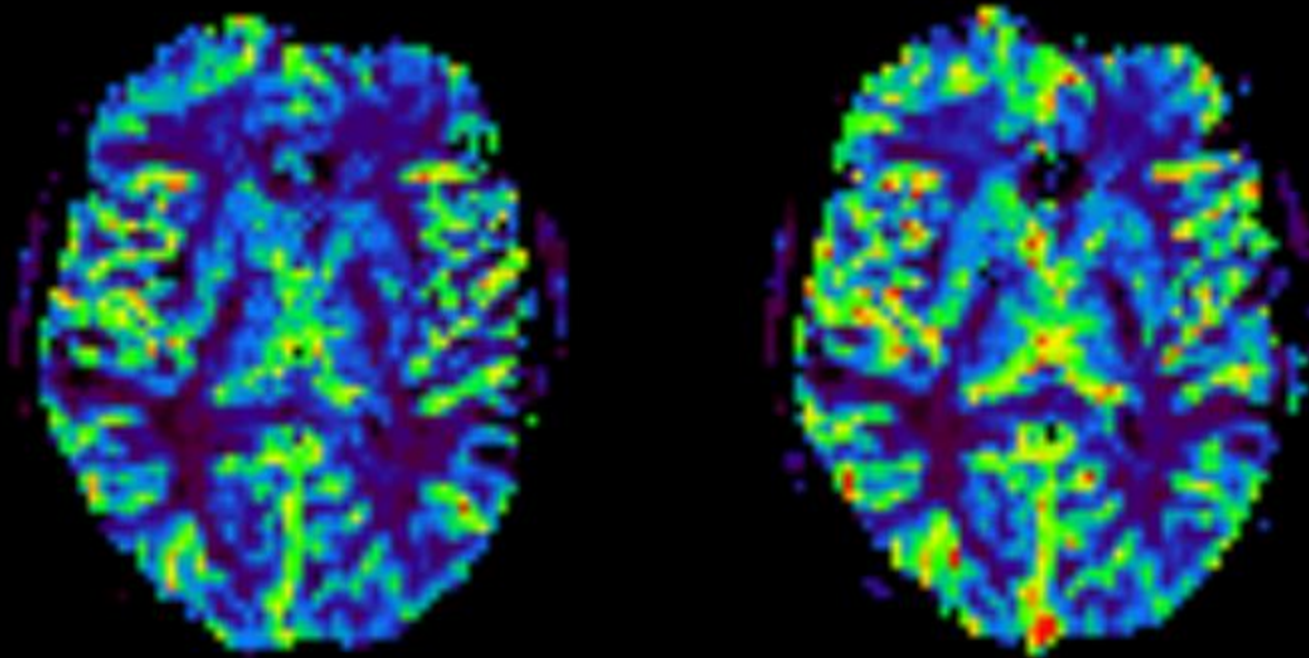
Findings (labeled)



Diagnostic Angiogram

Right M1 origin severe flow limiting stenosis with small adjacent collaterals

Findings: (labeled)



Cerebral Perfusion MRI Following Acetazolamide Challenge – Cerebral Blood Flow Map(CBF) Maps

Left: Pre-challenge, Right: Post-challenge MR perfusion Cerebral blood flow (CBF) maps show an augmentation of cerebral blood flow indicating the presence of cerebrovascular reserve.

Pertinent Labs

Factor VIII Activity 208, slightly Increased

Antithrombin III Activity

Beta 2 Glycoprotein IgG and IgM Antibodies

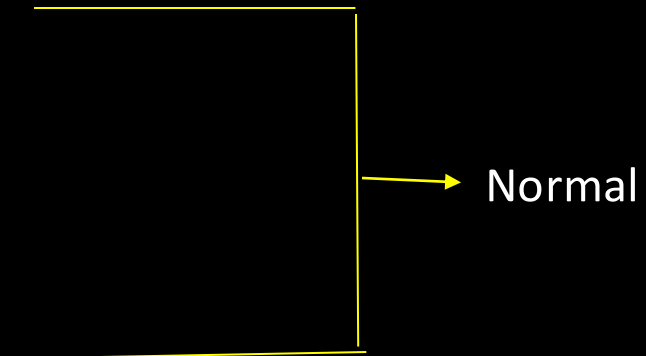
Lupus Screen DRVVT

Protein C Activity

Protein S Activity

Prothrombin G20210A

Other Lab findings are WNL



Final Dx:

Moyamoya Vasculopathy affecting Right anterior Circulation with
Acute ischemic right MCA stroke

Case Discussion

Etiology:

- Moyamoya disease is an idiopathic, progressive cerebrovascular disorder characterized by stenosis or occlusion of the internal carotid arteries and their main branches¹.
- About 10-15% of cases of moyamoya disease are familial, and some cases result from specific genetic mutations

The following types of moyamoya disease with the chromosome involved have been described in the literature²:

- MYMY1 (chromosome 3p)
- MYMY2 (*RNF213* gene on chromosome 17q25)
- MYMY3 (chromosome 8q23)
- MYMY4 (X-linked recessive condition characterized by moyamoya disease, short stature, hypergonadotropic hypogonadism, and facial dysmorphism)
- MYMY5 (*ACTA2* gene on chromosome 10q23)
- MYMY6 with achalasia (*GUCY1A3* gene on chromosome 4q32)

RNF213 gene is found in patients of Japanese and Korean descent and therefore moyamoya is more common in East Asian population

Presentation:

Patients usually present with TIA, ischemic/hemorrhagic stroke, seizure, or idiopathic and/or isolated stroke-like symptoms³

Pathophysiology :

Pathogenesis Of Moyamoya remains elusive. Authors report that that disease has multifactorial pathogenesis causing progressive narrowing of cerebral arteries. A **collateral circulation** develops around the blocked vessels to compensate for the blockage which resemble a “puff of smoke” (moyamoya in Japanese) on angiography, increasing the risk of ischemic and hemorrhagic strokes.

Diagnosis:

Cerebral angiography is the gold standard of diagnosing moyamoya disease and its progression⁴. According to Suzuki's system, it can be classified into six stages:

- Stage 1 Narrowing of carotid fork
- Stage 2 Initiation of the moyamoya and dilatation of intracranial main arteries
- Stage 3 Intensification of the moyamoya and defects of the anterior cerebral artery and middle cerebral artery
- Stage 4 Minimization of the moyamoya and defects of the posterior cerebral artery
- Stage 5 Reduction of the moyamoya and development of external carotid artery collaterals
- Stage 6 Disappearance of the moyamoya and circulation only via external carotid artery and vertebral artery

Magnetic resonance imaging (MRI) typically serves as an initial diagnostic tool due to its high sensitivity and noninvasive nature notable radiographic feature, the "ivy sign," reflects increased signal intensity on T2-FLAIR and appears in nearly half of pediatric patients with Suzuki stage III or IV disease⁵

Magnetic resonance angiography (MRA) provides information on cerebral arteries and the degree of narrowing

Genetic Testing

Test Performed: Hereditary Moyamoya Disease Panel

Genetic testing revealed a variant of uncertain significance (VUS) in the RNF213 gene. While the RNF213 gene is associated with autosomal dominant and recessive Moyamoya disease type 2, this specific variant is present in the general population and has not been reported in individuals with Moyamoya disease.

Subsequently, Parental testing for this variant was recommended to determine if it was inherited from an unaffected parent or de novo (new) .

Management:

- Acute therapy for strokes or intracranial bleeding is performed as per standard protocols⁶.
- Conservative management is directed towards maintaining cerebral blood flow and preventing further ischemic and hemorrhagic events. Surgical intervention, is of utmost importance as medical therapies act only as secondary prevention such as antiplatelet drugs for clot prevention and do not halt disease progression.

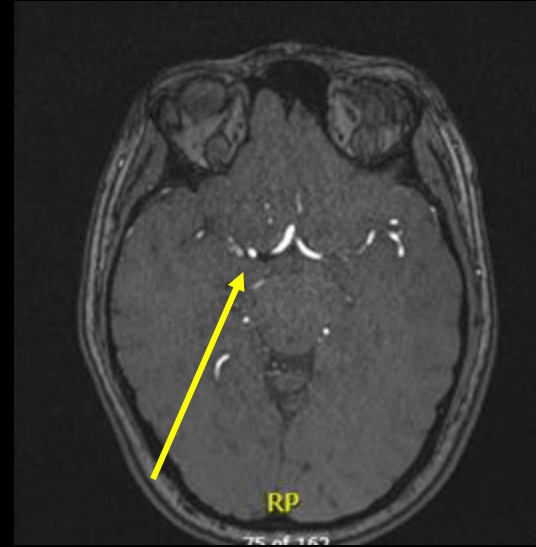


Indirect Revascularization

- **EDAS**
(encephaloduroarteriosynangiosis)
- **EMS**
(encephalomyosynangiosis)

Direct Revascularization

- **STA-MCA** procedure



Follow up MRA WO IVCon 2025

Redemonstrated severe right M1 segment stenosis with associated lenticulostriate collaterals and faint distal M1 level reconstitution

References:

1. **Kuroda, S., & Houkin, K.** (2008). Moyamoya disease: current concepts and future perspectives. *The Lancet Neurology*, 7(11), 1056–1066. [https://doi.org/10.1016/S1474-4422\(08\)70240-0](https://doi.org/10.1016/S1474-4422(08)70240-0)
2. **Scott, R. M., & Smith, E. R.** (2009). Moyamoya disease and Moyamoya syndrome. *New England Journal of Medicine*, 360(12), 1226–1237. <https://doi.org/10.1056/NEJMra0804622>
1. **Chiu, D., Shedden, P., Bratina, P., & Grotta, J. C.** (1998). Clinical features of Moyamoya disease in the United States. *Stroke*, 29(7), 1347–1351. <https://doi.org/10.1161/01.STR.29.7.1347>
2. Uchino, K., & Kudo, S. (2014). Neuroimaging diagnosis and collateral circulation in Moyamoya disease. *Neurologia Medico-Chirurgica*, 54(9), 694–705.: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4031769/>
3. Kim, J. H., & Kim, J. E. (2019). Imaging of Moyamoya disease and Moyamoya syndrome: Current status. *Journal of Stroke*, 21(1), 42–52. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6426357/>
1. **Powers, W. J., Rabinstein, A. A., Ackerson, T., et al.** (2019). Guidelines for the early management of patients with acute ischemic stroke: 2019 update to the 2018 guidelines. *Stroke*, 50(12), e344–e418. <https://doi.org/10.1161/STR.0000000000000211>