Diagnostic Slide Session
AANP 2018

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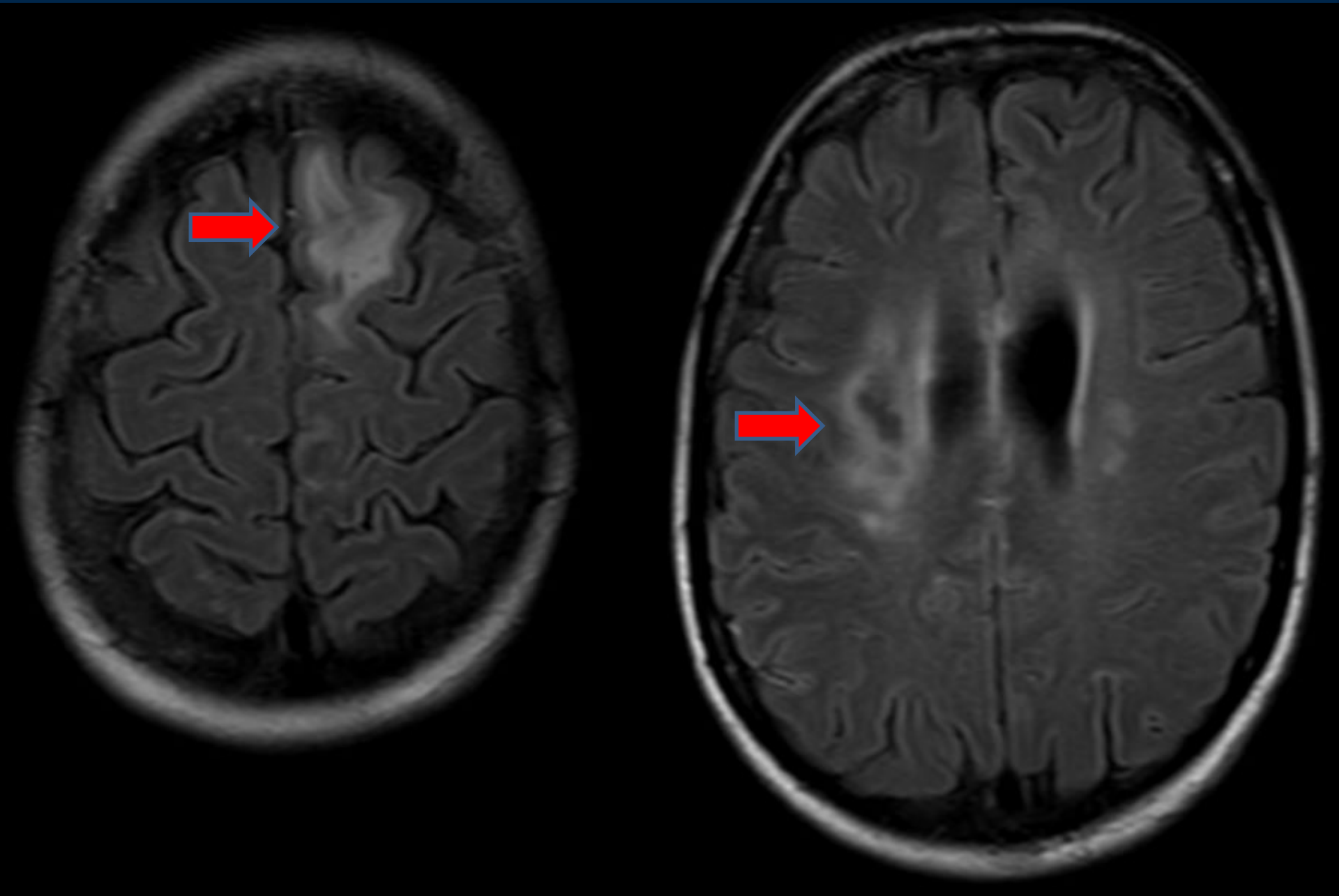
No financial disclosures

50 year old female presenting with an acute episode of expressive dysphasia and right-sided weakness

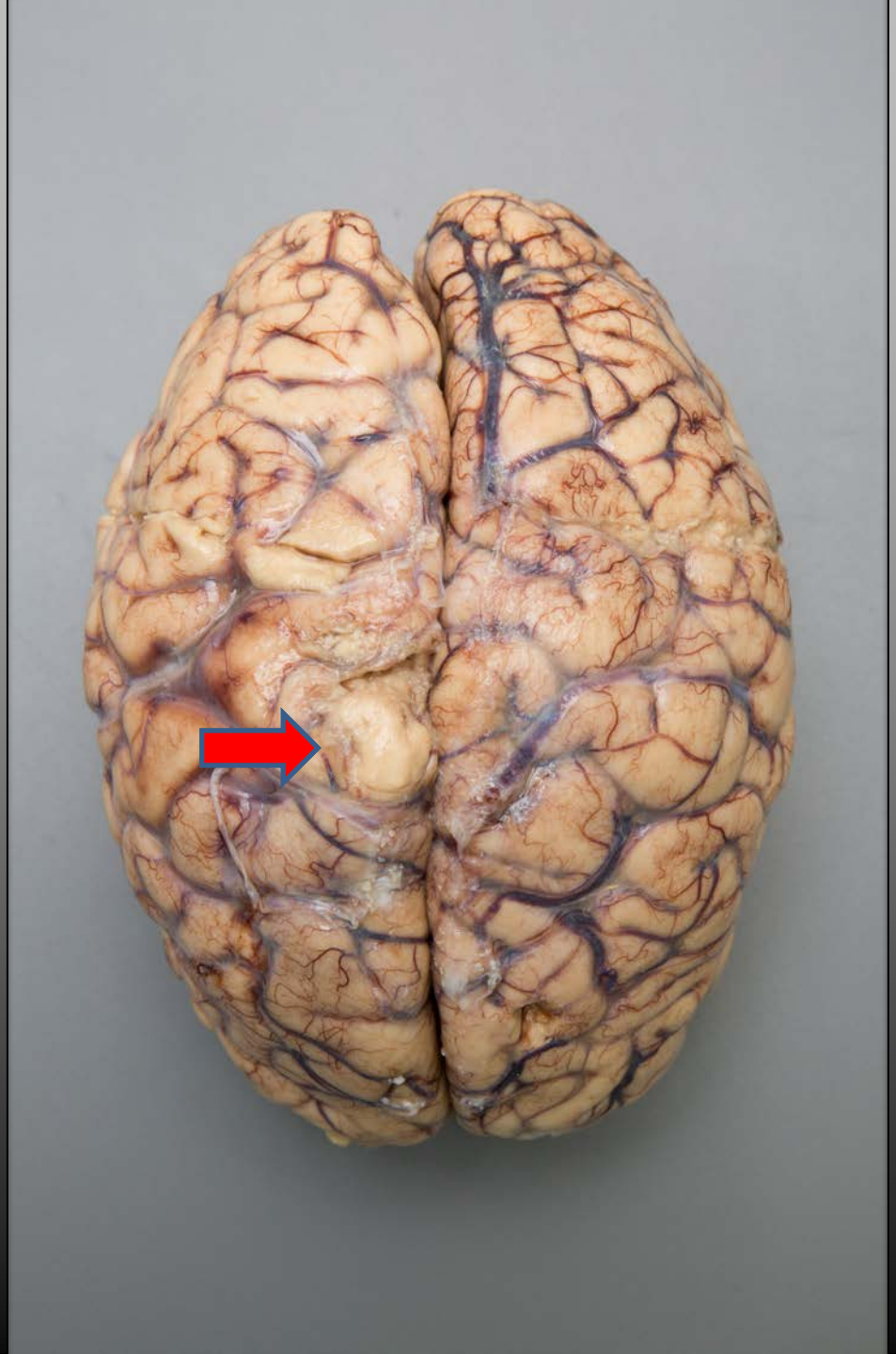


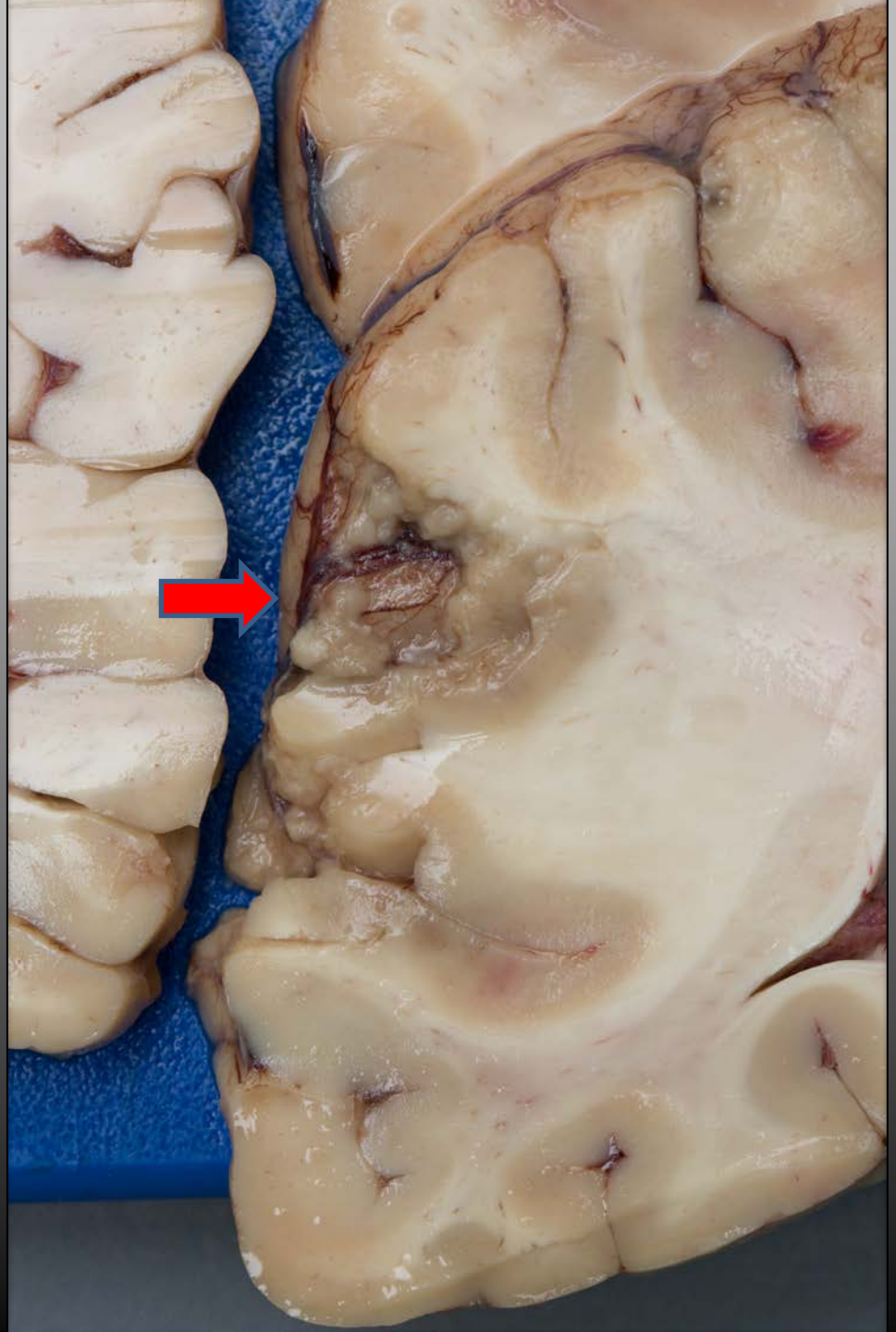
Clinical History

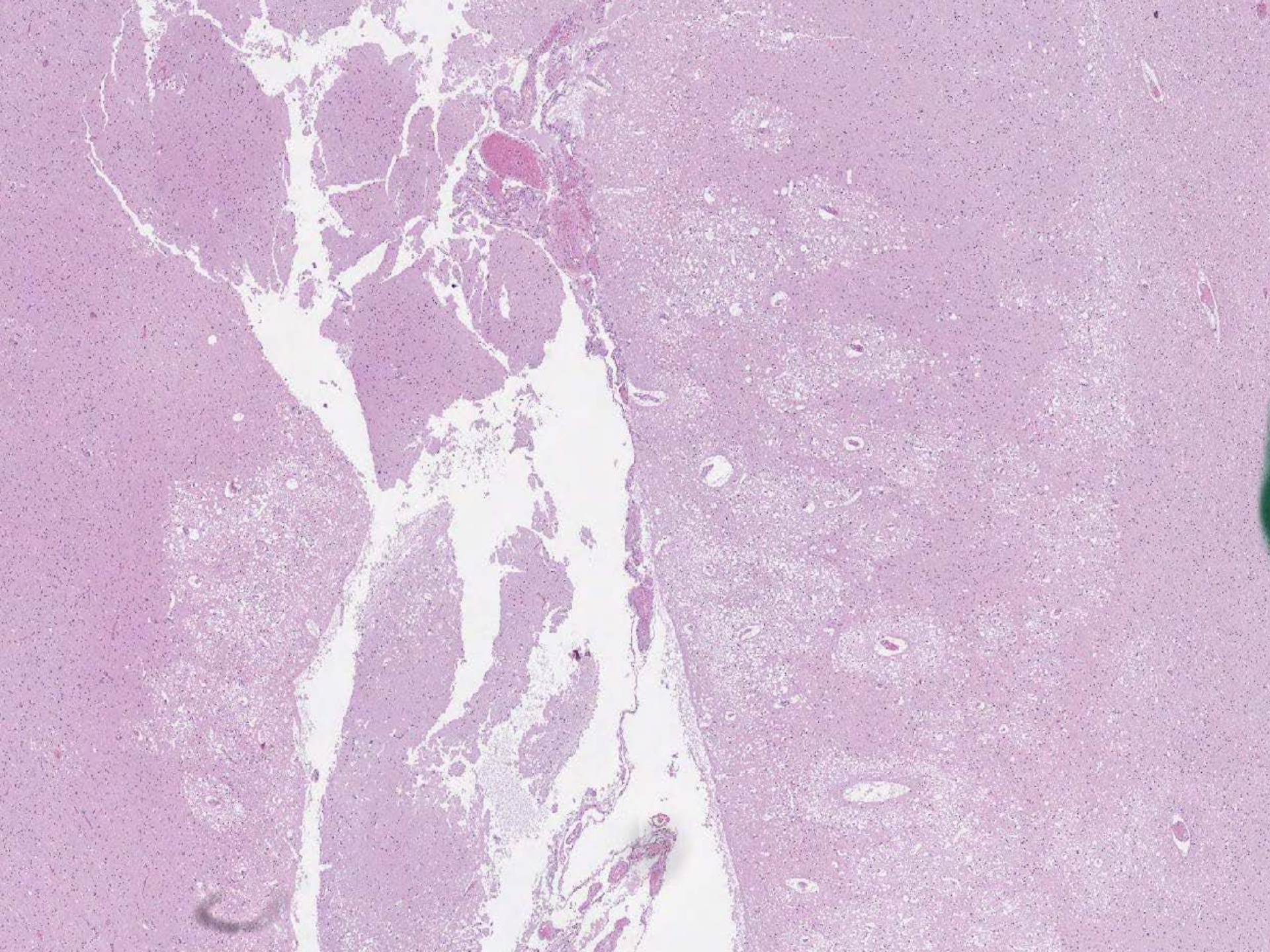
- Frontal lobe biopsy showed only reactive gliosis
- Symptoms improved with steroid treatment
- Experienced multiple acute events over several years with worsening focal deficits
- Became unresponsive after an acute event clinically believed to be an ischemic stroke, and the family withdrew care

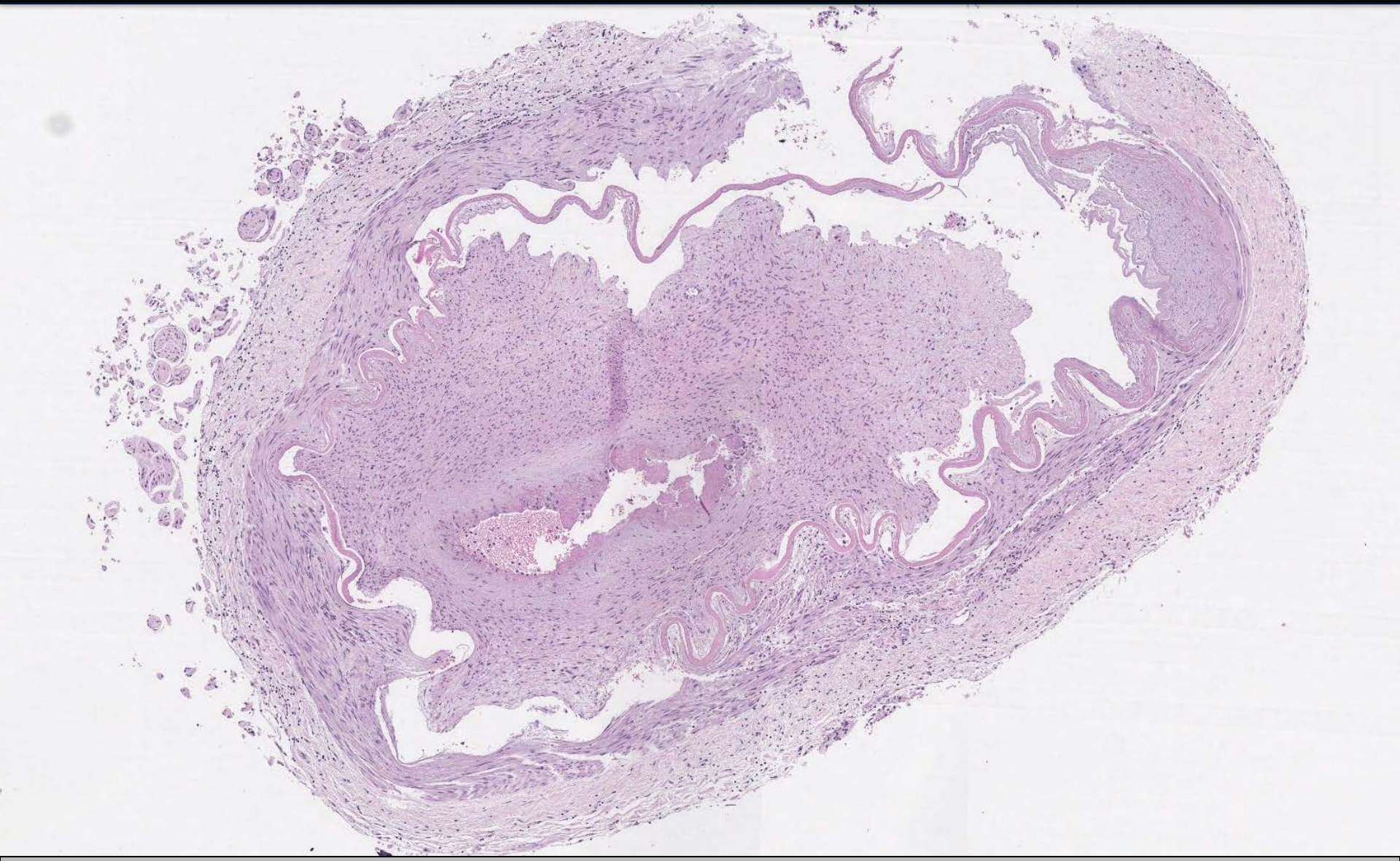
Autopsy gross findings

- Brain weight: 1450 grams
- Multiple areas of softening in the left frontal, right fronto-parietal, right superior parietal, left mid-parietal, and left occipital lobes
- Markedly narrowed lumen of internal carotid arteries



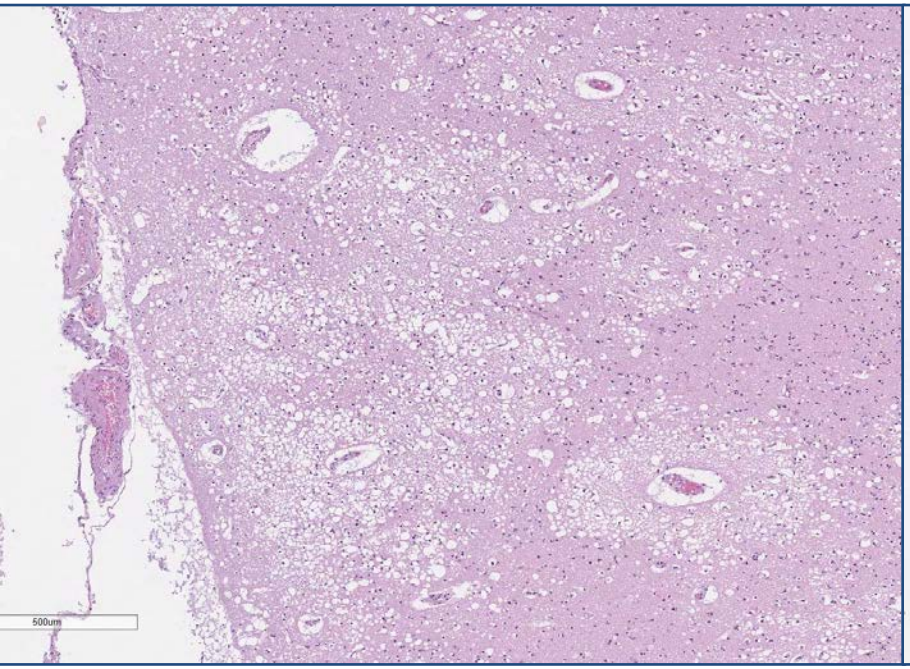


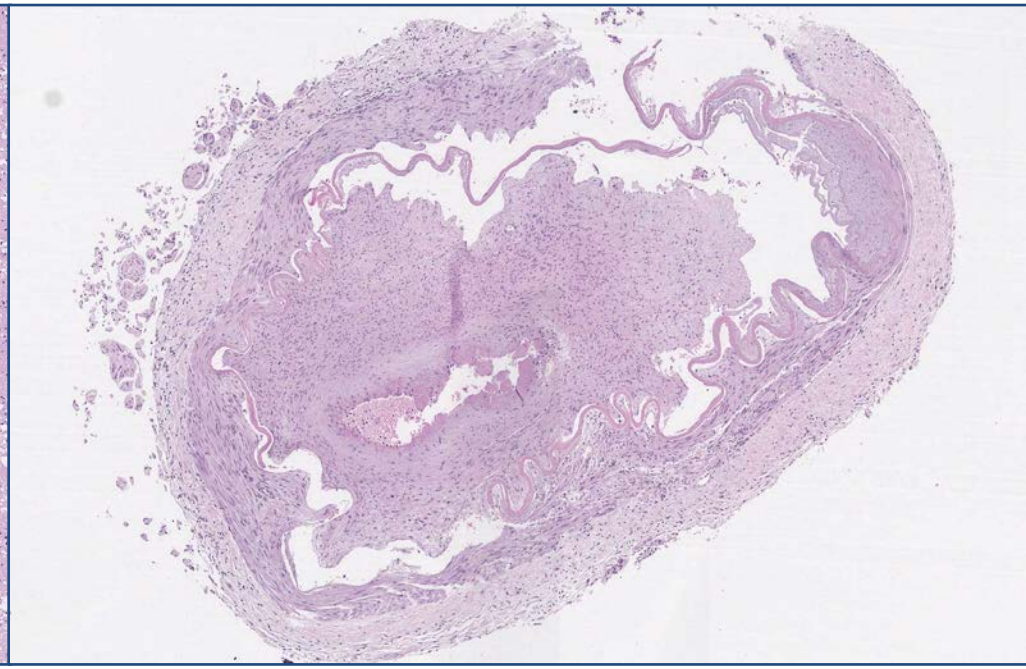




Discussion

- 50 year old woman with episodically worsening focal deficits and multifocal gray-white lesions on imaging
- Multiple areas of softening in the left frontal, right fronto-parietal, right superior parietal, left mid parietal, and left occipital lobes





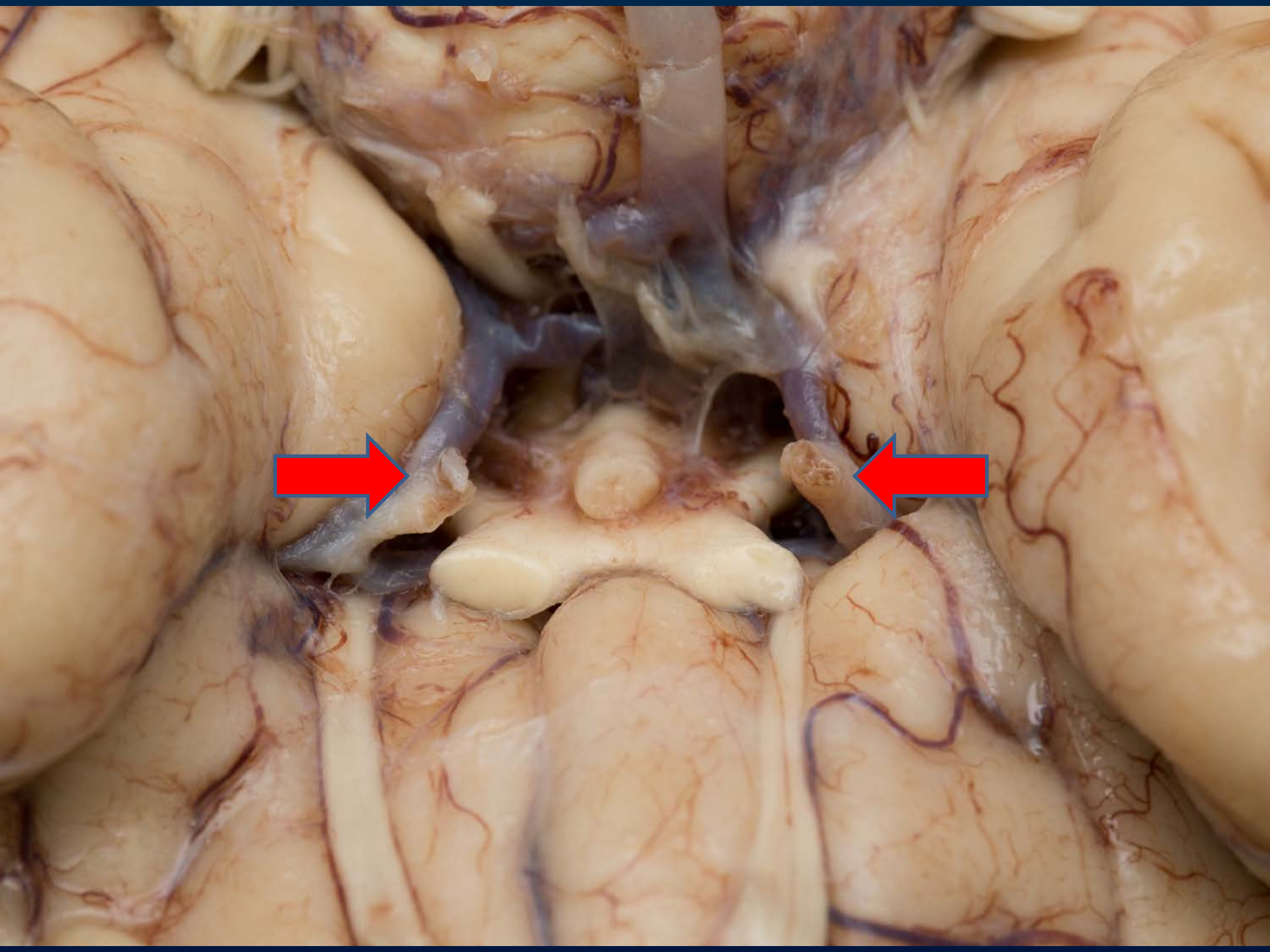
Additional findings

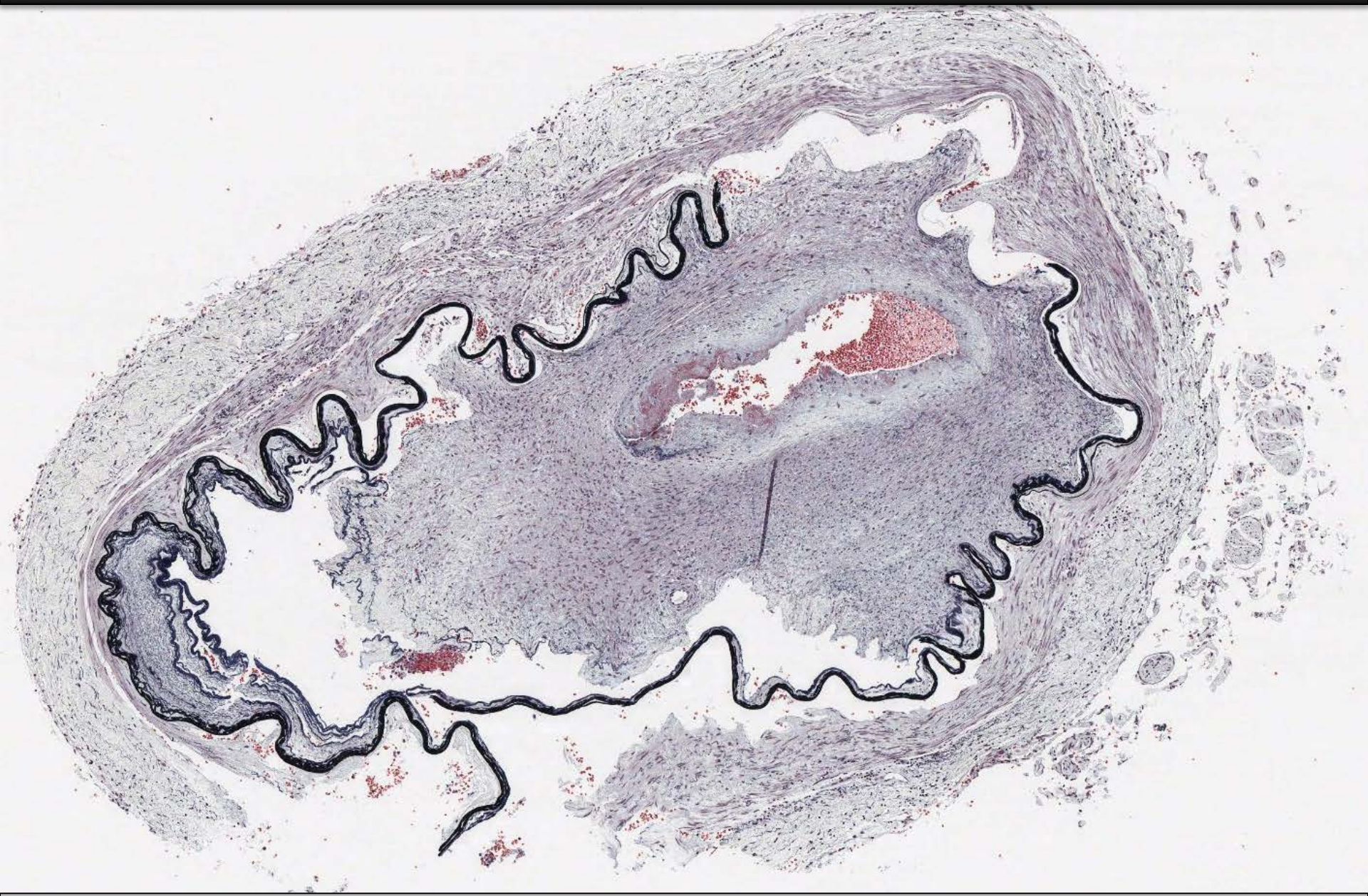
Clinical

- Angiography showing focal narrowing of the internal carotid, celiac, renal, iliac, and femoral artery
- Negative workup for autoimmune disease or vasculitis

Autopsy

- Watershed distribution of cortical lesions
- No inflammation in cortical sections
- No inflammation or giant cells in carotid artery





Differential Diagnosis

Diagnosis	Characteristics
Demyelinating disease	• Disease limited to white matter • Should not have vascular distribution
Atherosclerotic disease	• Patients with risk factors • Vascular lesions distributed to areas of turbulent flow
Dissection / aneurysm	• Identified by radiologic or pathologic changes • Dissection and aneurysm may be consequence of fibromuscular dysplasia
Vasculitis	• Systemic symptoms, anemia, thrombocytopenia, or elevated acute-phase reactants • Pathologic evidence of inflammation
Fibromuscular dysplasia	• “String of beads” appearance on angiography • Intimal or medial proliferation on pathology

Final Diagnosis

Fibromuscular dysplasia (FMD), intimal type

Fibromuscular Dysplasia

- Noninflammatory, nonatherosclerotic vasculopathy
- Mean age: 52 years, 90% of adults are women
- Etiology unknown:
  - Genetic associations (*PHACTR1* gene)
  - Hormonal associations
- Most cases are diagnosed by angiography

Classification

	Focal		Multifocal
Radiographic Findings	Circumferential or tubular stenosis		String of beads
Clinical	Younger patients with renal disease and hypertension		Older patients
Histologic classification	Intimal	Adventitial	Medial
Histologic Findings	Intimal collagen deposition	Collagen extending into periarterial fat	Alternating with absent lamina in dilated areas
Incidence	~10%	<1%	>90%

Clinical characteristics

Sites of involvement

- Renal (80%)
- Extracranial carotid (74%)
- Vertebral (37%)
- Mesenteric (26%)
- Intracranial carotid (17%)

Presentation

- Hypertension (64%)
- Headache (52%)
- Pulsatile tinnitus (28%)
- Dizziness (26%)
- Carotid bruit (22%)

Olin JW, Froehlich J, Gu X, Bacharach JM, Eagle K, Gray BH, Jaff MR, Kim ES, Mace P, Matsumoto AH, McBane RD, Kline-Rogers E, White CJ, Gornik HL. The United States Registry for Fibromuscular Dysplasia: results in the first 447 patients. Circulation. 2012;125:3182–3190.

Conclusion

- Consider FMD in the differential when multifocal CNS lesions are present
- CNS circulation is a common site of involvement
- Pathology may be necessary to resolve the differential

References

- Olin JW, Froehlich J, Gu X, Bacharach JM, Eagle K, Gray BH, Jaff MR, Kim ES, Mace P, Matsumoto AH, McBane RD, Kline-Rogers E, White CJ, Gornik HL. The United States Registry for Fibromuscular Dysplasia: results in the first 447 patients. Circulation. 2012;125:3182–3190.

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- Savard S, Steichen O, Azarine A, Azizi M, Jeunemaitre X, Plouin PF. Association between 2 angiographic subtypes of renal artery fibromuscular dysplasia and clinical characteristics. Circulation 2012; 126:3062–3069.

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