MAYO CLINIC

61st Annual Diagnostic Slide Session 2020

Case 10

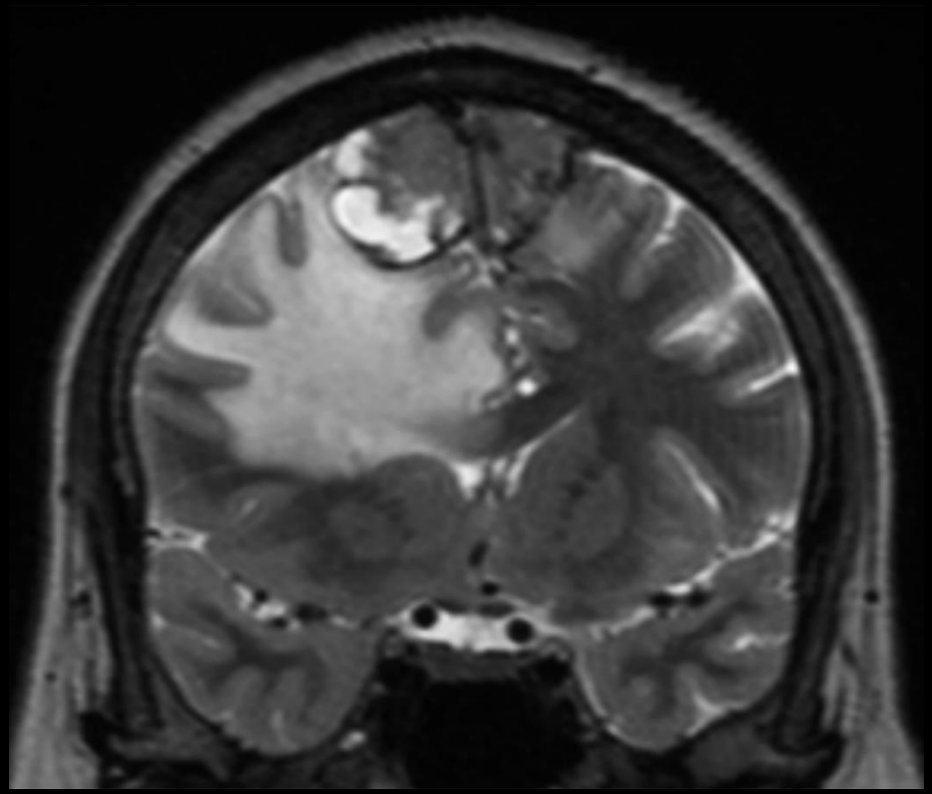
M. Adelita Vizcaino M.D.

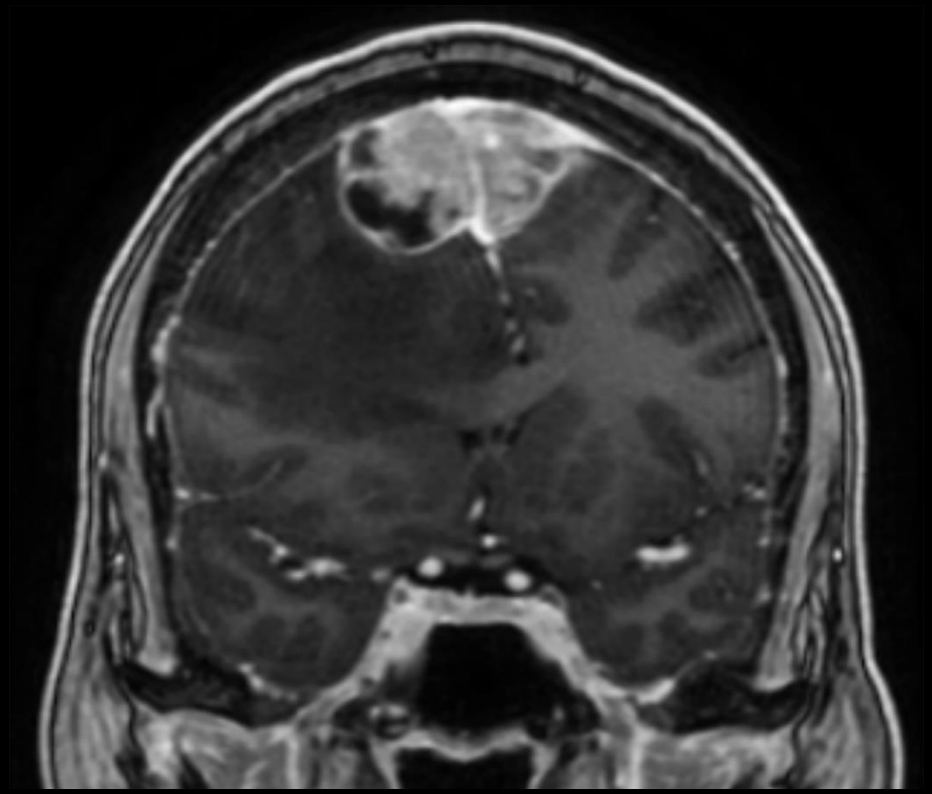
Department of Laboratory Medicine and Pathology
Mayo Clinic Rochester, MN

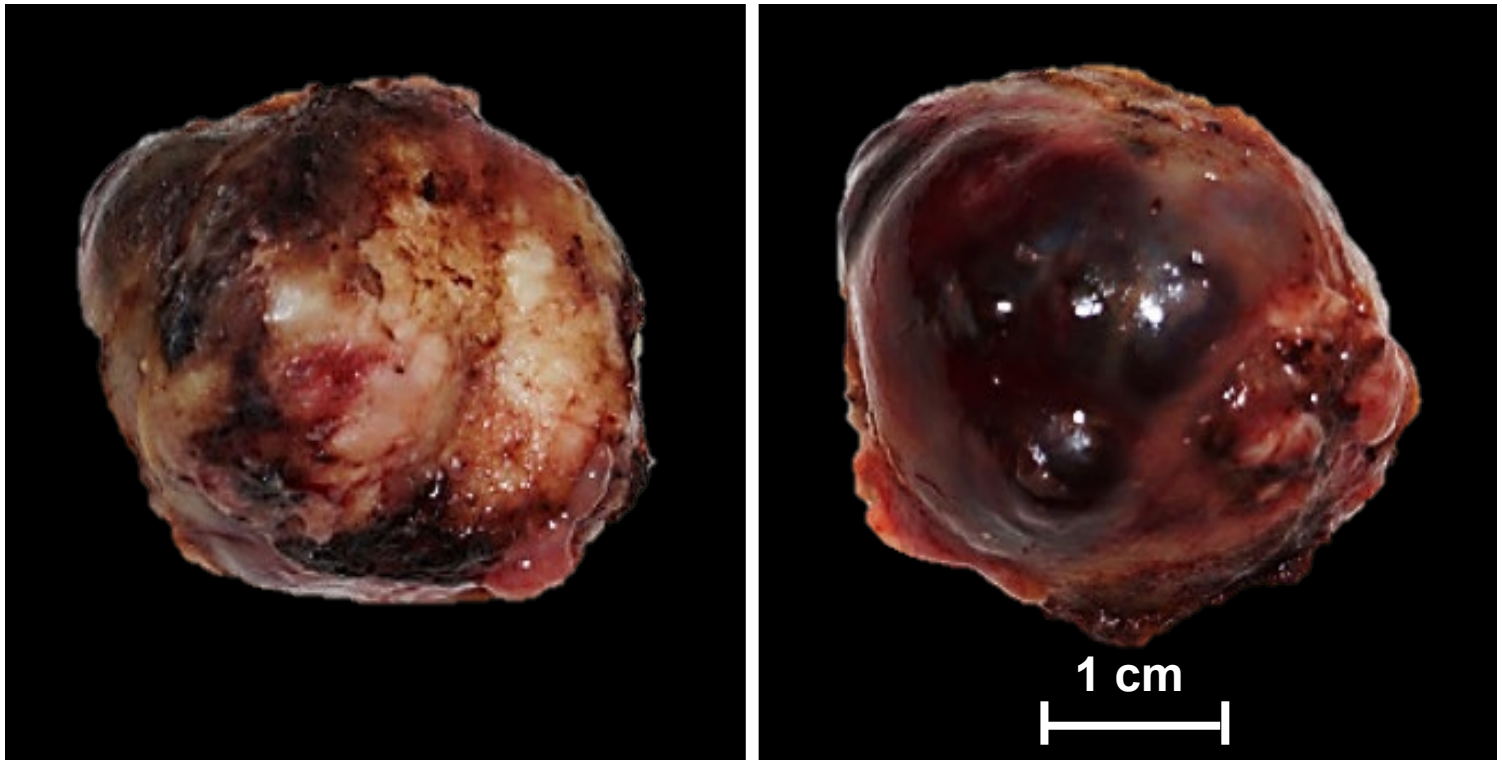
No financial disclosures

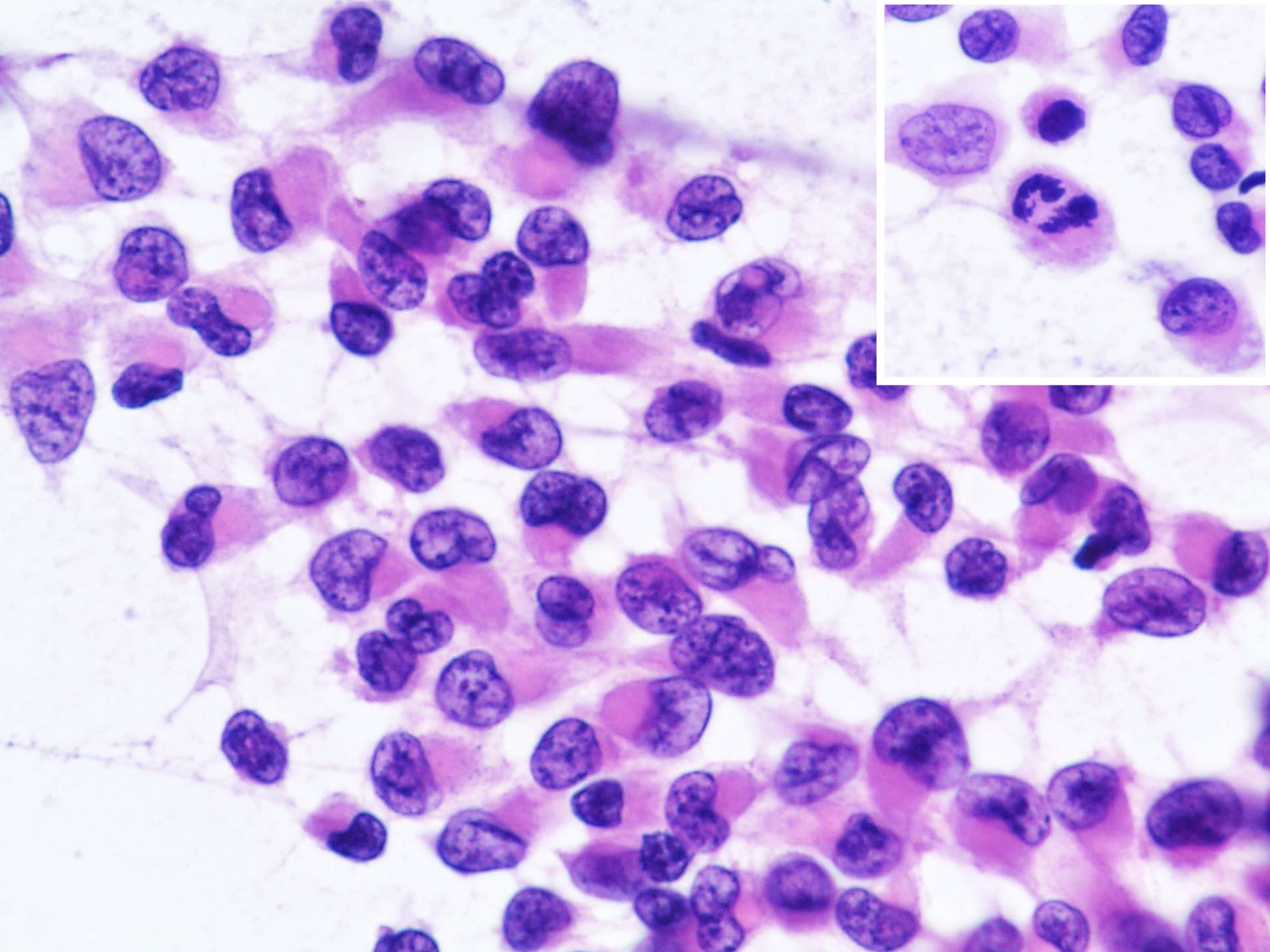
Case 10
Clinical summary

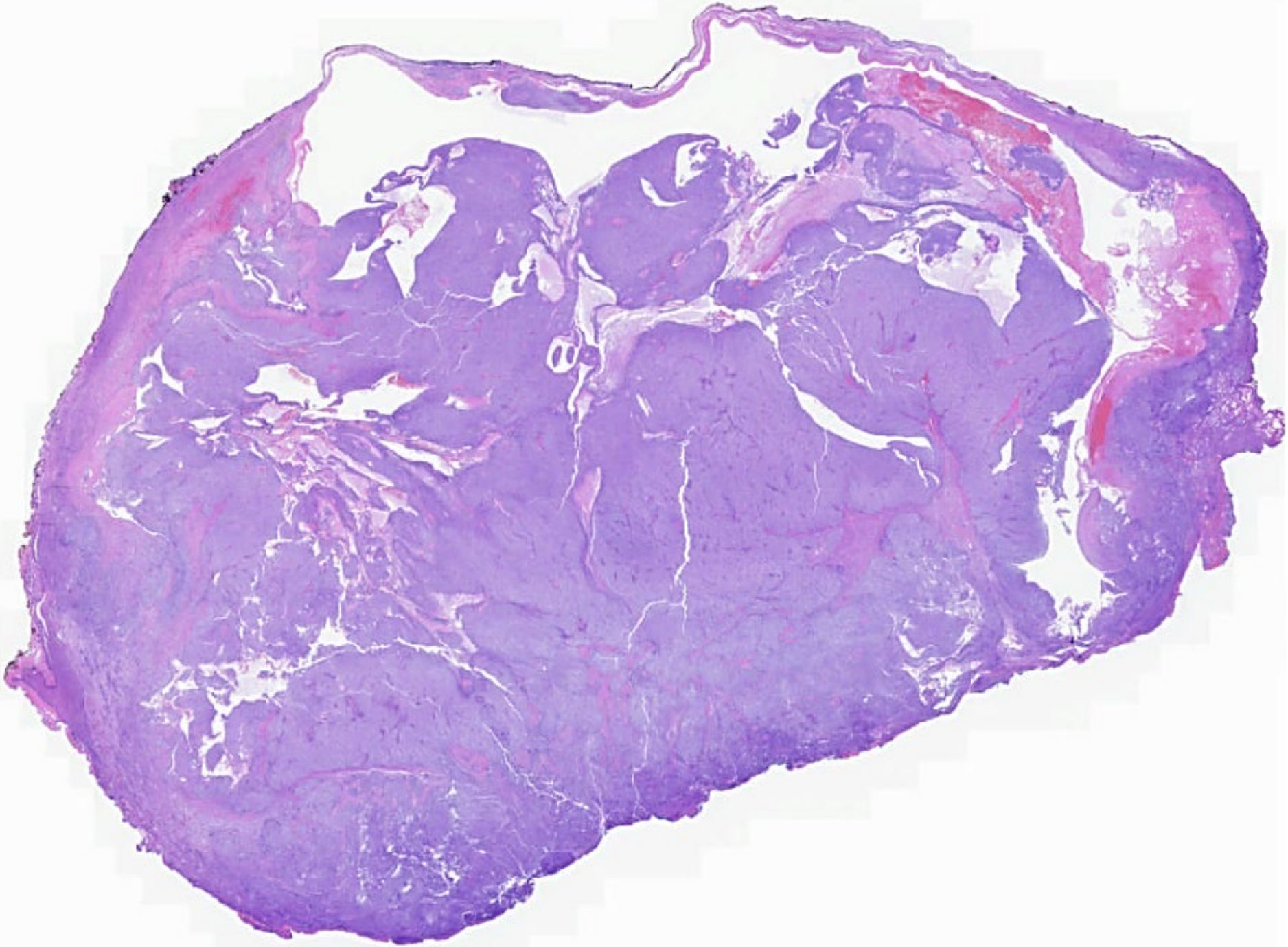
- 50-year-old woman
  - Several falls due to syncope with loss of consciousness

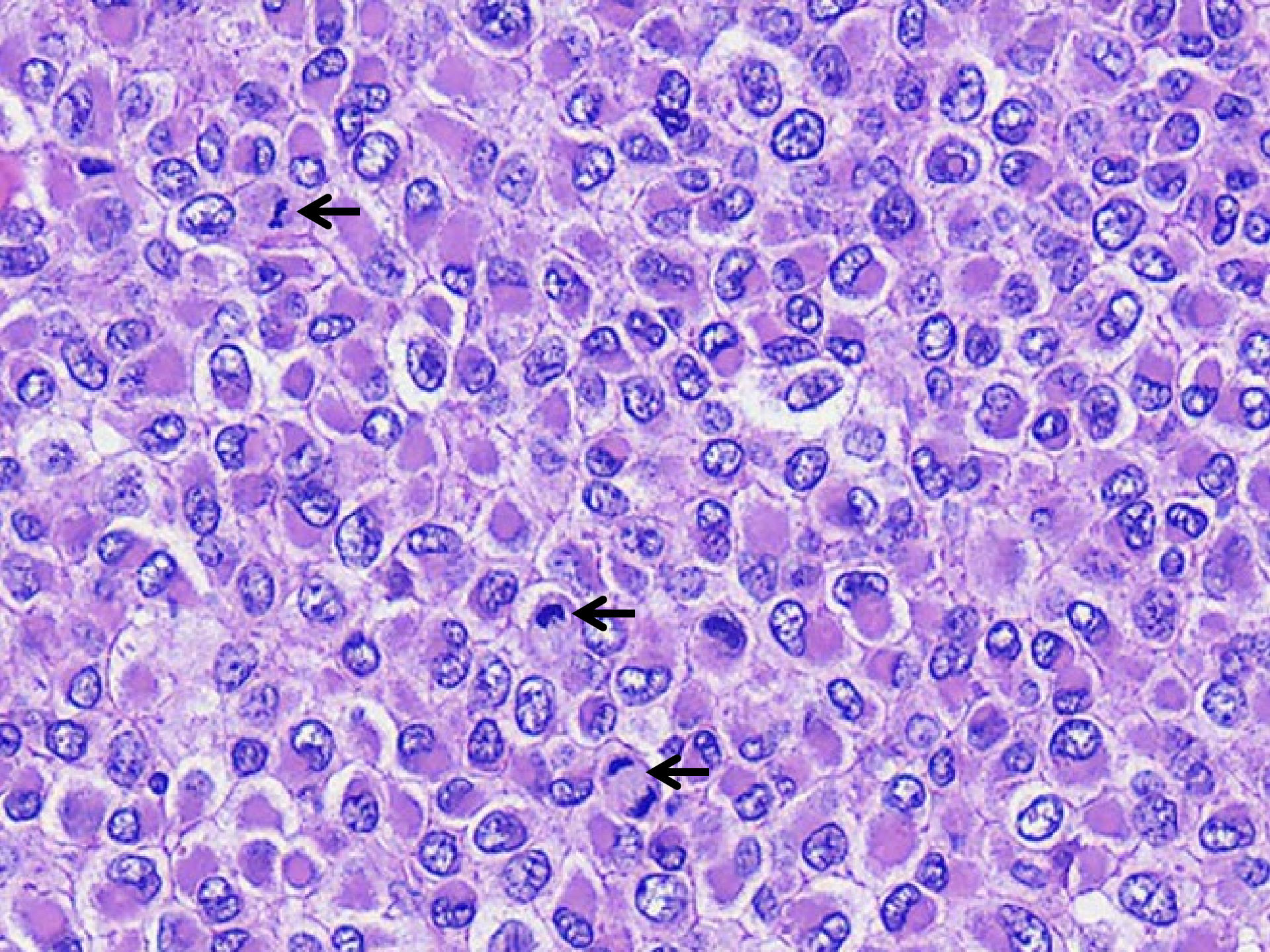




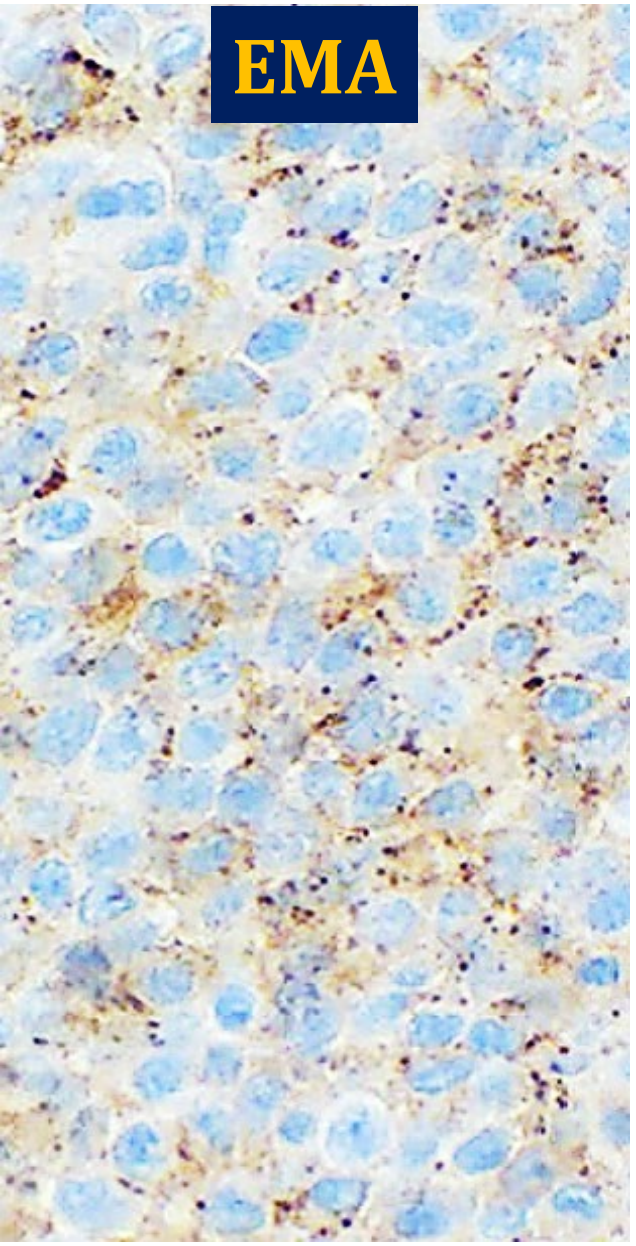

1 cm

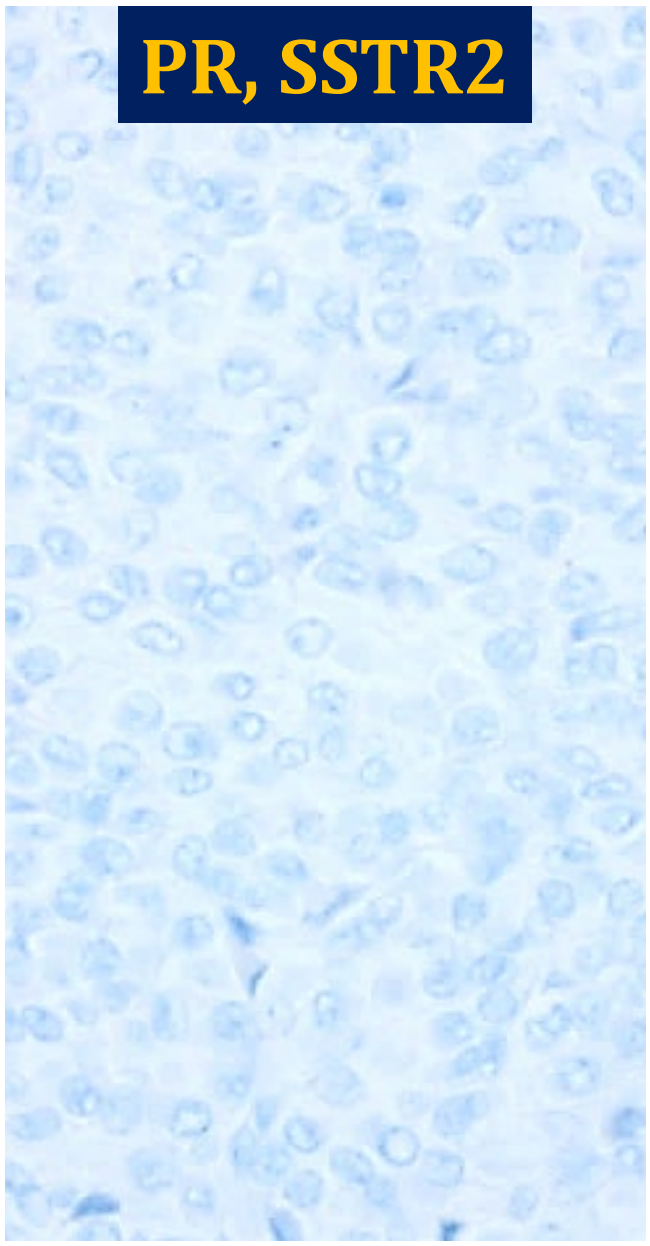


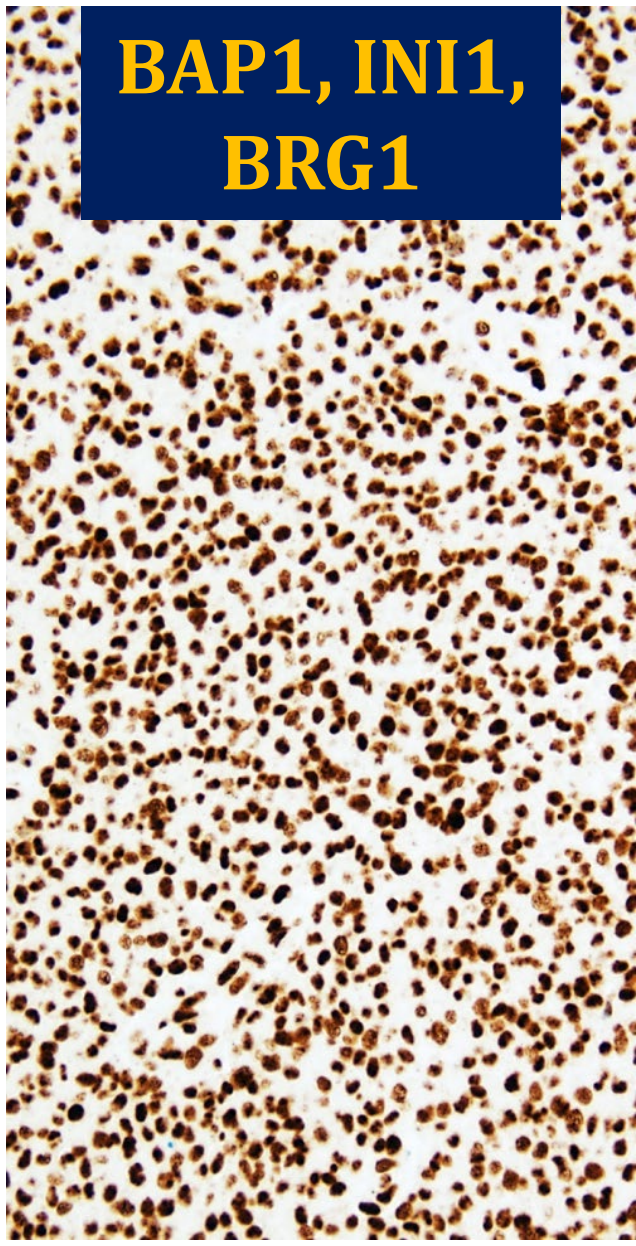


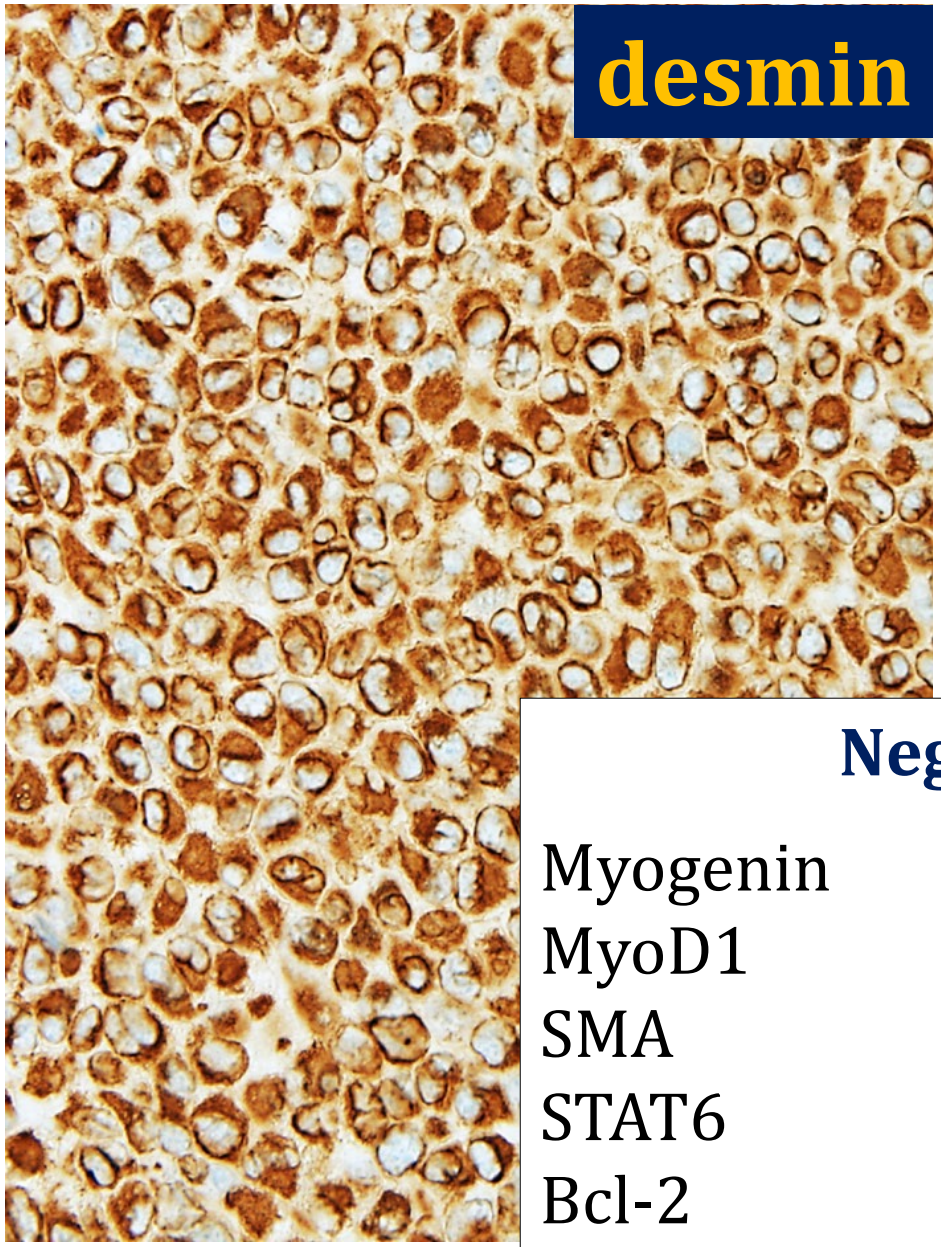


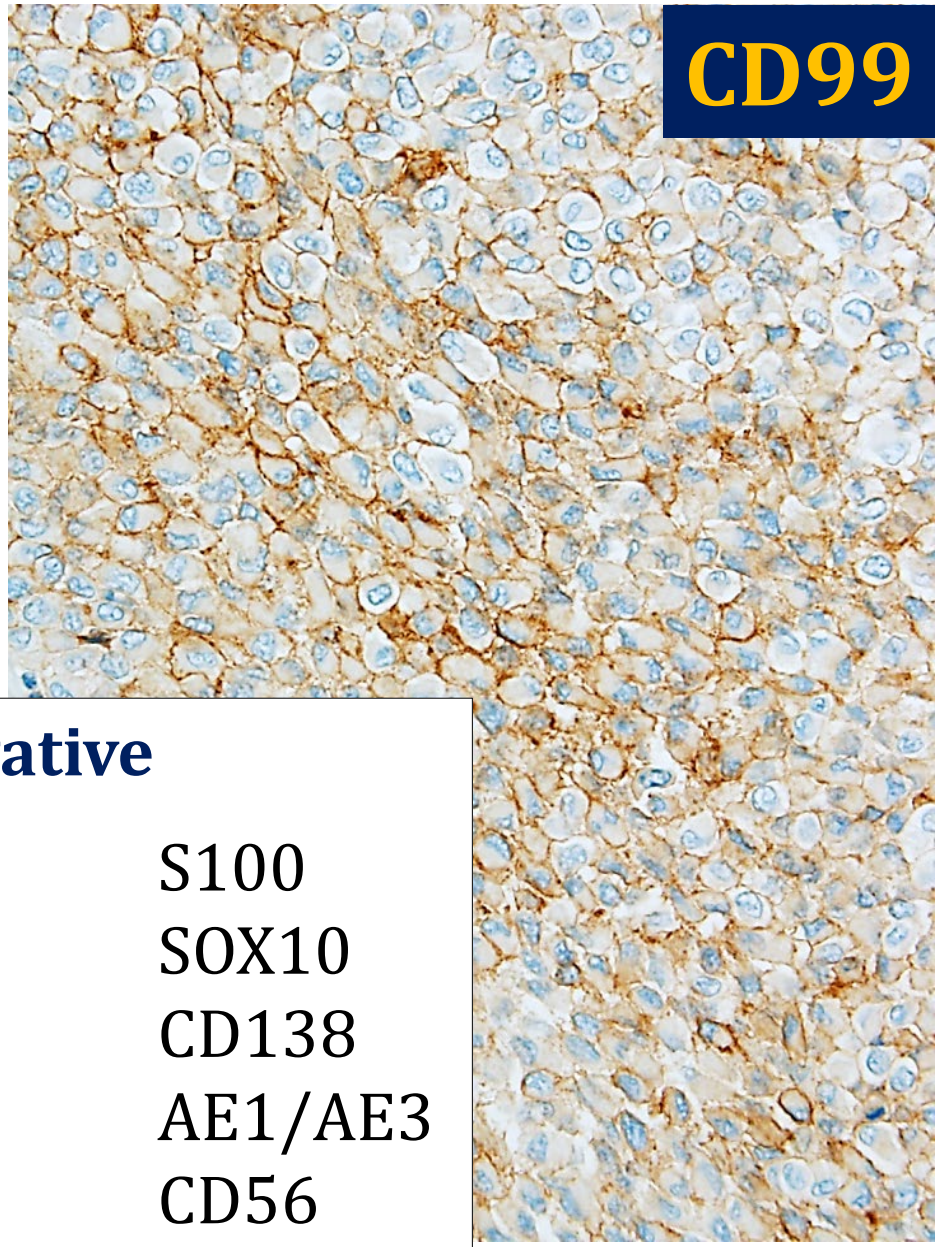
Diagnosis?


EMA


PR, SSTR2


BAP1, INI1,
BRG1

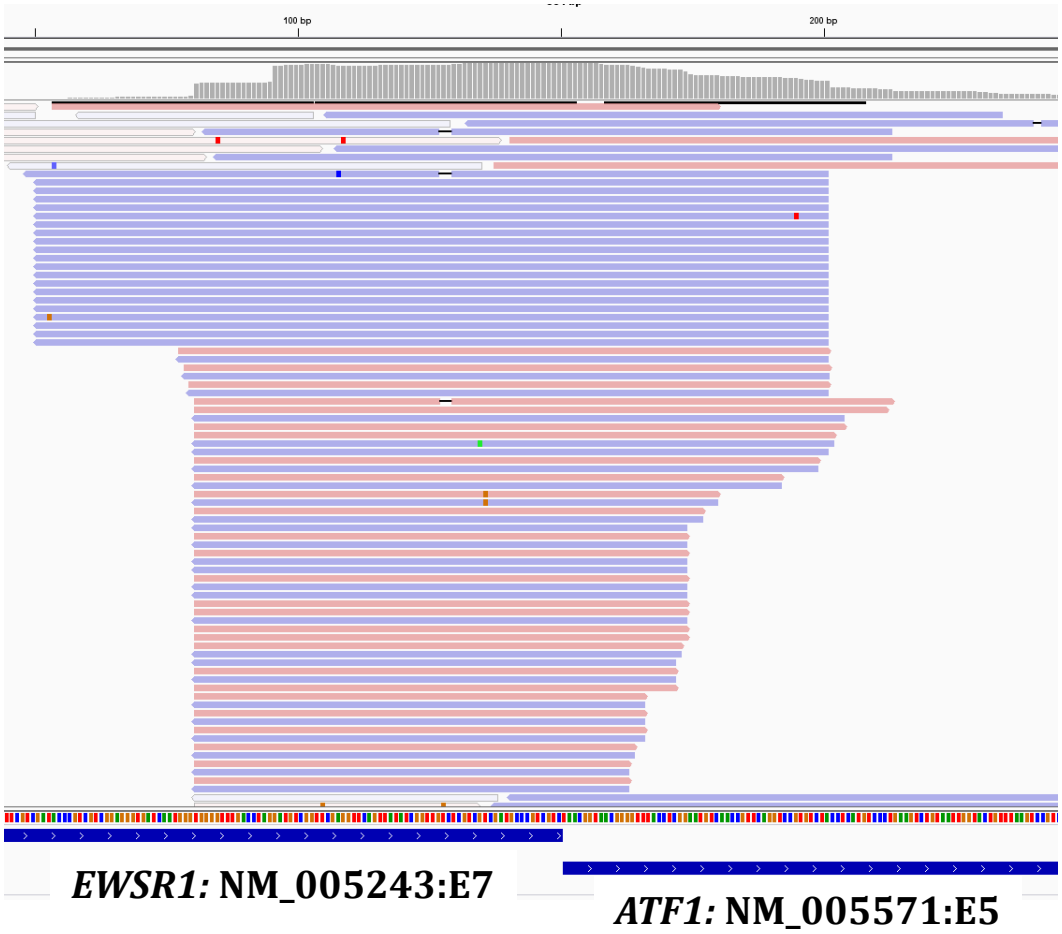




Negative

Myogenin	S100
MyoD1	SOX10
SMA	CD138
STAT6	AE1/AE3
Bcl-2	CD56
CD34	

Sarcoma Targeted Gene Fusion Panel
EWSR1-ATF1 fusion

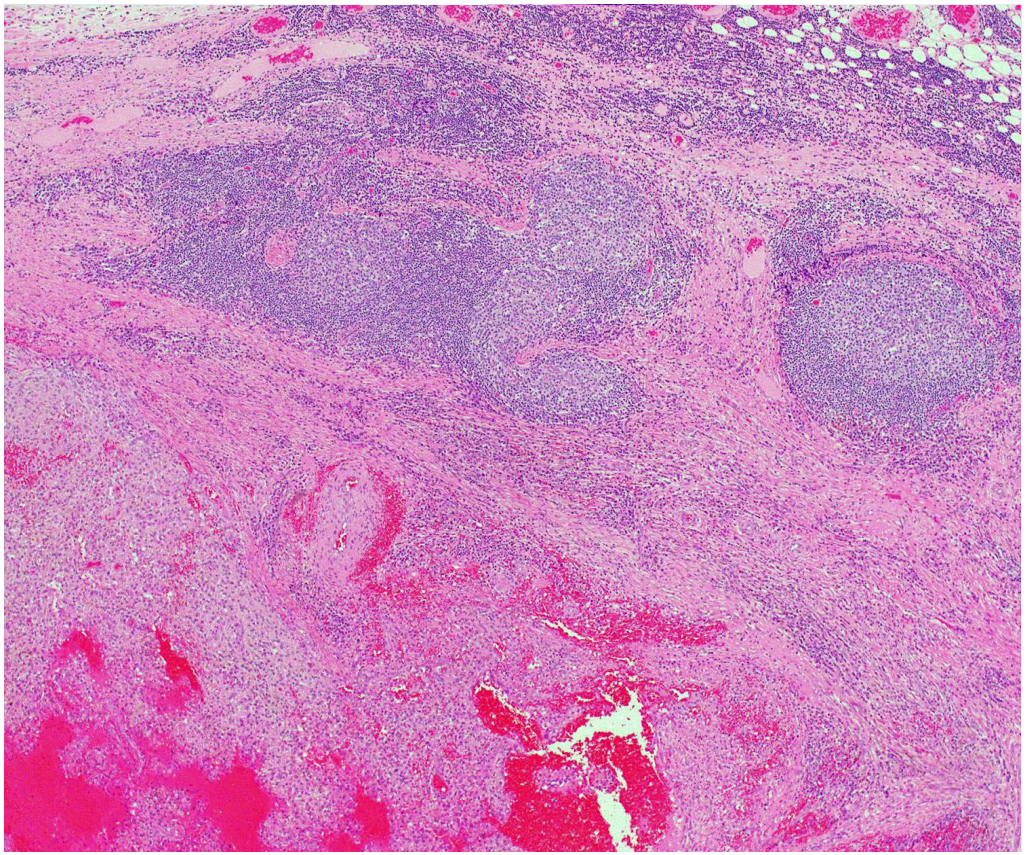


Diagnosis:

Angiomatoid fibrous histiocytoma with rhabdoid features

Angiomatoid fibrous histiocytoma (AFH)

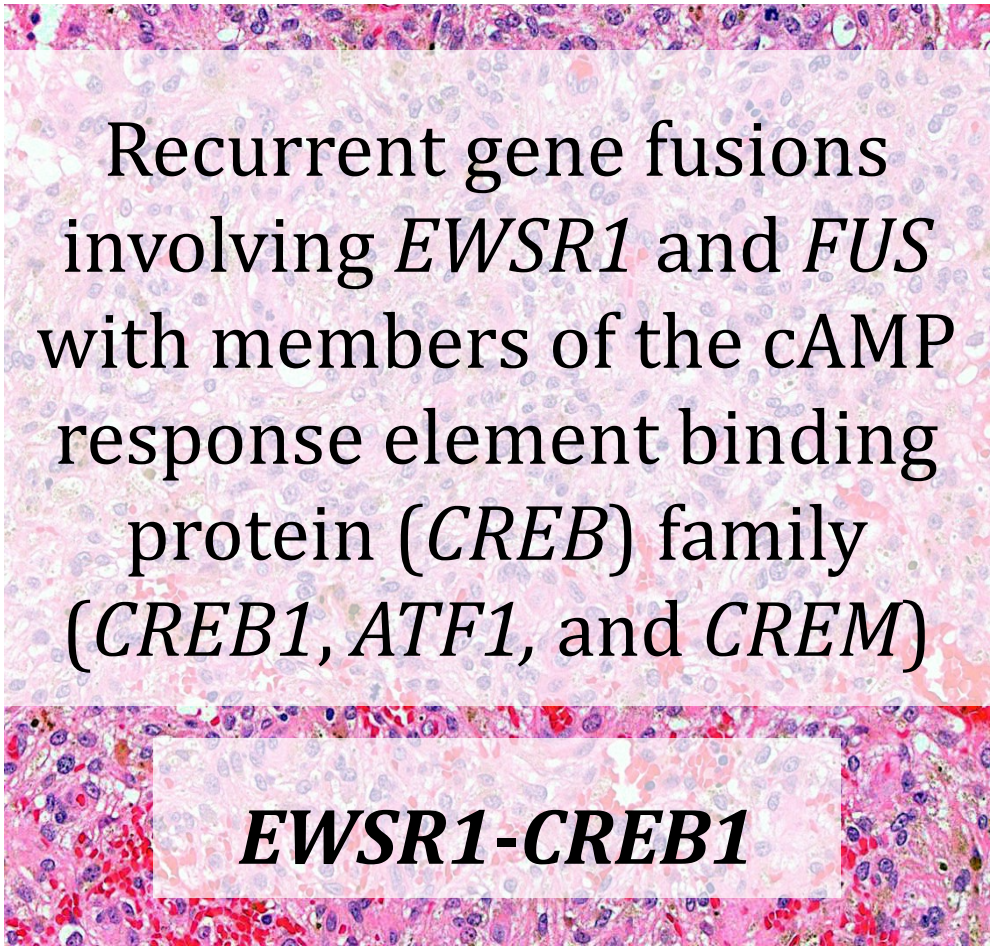
- Rare, primarily in the extremities of young people
- Encapsulated, with hemorrhagic pseudocystic spaces lacking endothelium
- Variable lymphoplasmocytic inflammation



Thway K et al. Arch Pathol Lab Med. 2015
Chen G et al. Mod Pathol. 2011
Costa MJ et al. Am J Surg Pathol. 1990

Angiomatoid fibrous histiocytoma (AFH)

- Histiocyte-like cells
- Rhabdoid features uncommon
- Variable EMA, desmin, SMA, CD68, and CD99 expression



Konstantinidis A et al. World Neurosurg. 2019
Ghanbari N et al. Cureus 2019
Alshareef M et al. World Neurosurg. 2016

Intracranial AFH

- Usually in children and young adults
- Intra or extra-axial
- Prominent myxoid features (uncommon in extracranial AFH)
- Overlap with intracranial myxoid mesenchymal tumor
  - AFH-like tumors

Konstantinidis A et al. World Neurosurg. 2019
Gareton A et al. J Neuropathol Exp Neurol. 2018
Dunham C et al. Am J Surg Pathol. 2008

Intracranial myxoid mesenchymal tumor (MMT)

- Recently described entity
- Variable myxoid background
- Resembles the myxoid variant of AFH
- Shares genetic alterations with AFH (frequent fusions of *EWSR1* and *CREB* family genes)

Kao YC et al. Am J Surg Pathol. 2017
Bale et al. Brain Pathol. 2018
Ballester L et al. J Neuropathol Ex Neurol. 2020

IC AFH, AFH-like and MMT

Case #	Reference	Age/Sex	Location	Diagnosis	Molecular fusion
1	Dunham et al. (2008)	25/M	Occipital	AFH	*EWSR1-ATF1*
2	Ochalski et al. (2010)	35/M	Temporal	AFH	*EWSR1* rearrangement
3	Hansen et al. (2015)	17/F	Extra-axial (occipital)	AFH	*EWSR1* rearrangement
4	Alshareef et al. (2016)	58/F	CPA/middle cranial fossa	AFH	*EWSR1* rearrangement
5	Kao et al. (2017)	15/F	Meningeal	MMT	*EWSR1-CREM*
6	Kao et al. (2017)	23/F	Meningeal (occipital)	MMT	*EWSR1-CREB1*
7	Kao et al. (2017)	20/M	Parenchymal (frontal)	MMT	*EWSR1-CREB1*
8	Kao et al. (2017)	12/M	Parenchymal (frontal)	MMT	*EWSR1-ATF1*
9	Spatz et al. (2018)	22/F	Right occipital	AFH	Not assessed
10	Gareton et al. (2018)	19/M	Intra and extra-axial (CPA)	AFH-like	*EWSR1-CREM*
11	Sciot et al. (2018)	17/F	Parenchymal (frontal)	MMT	*EWSR1-ATF1*
12	Bale et al. (2018)	12/M	Abutting dura (posterior fossa/cerebellar)	MMT	*EWSR1-CREB1*
13	Bale et al. (2018)	14/F	Intraventricular (left lateral)	MMT	*EWSR1-CREB1*
14	Bale et al. (2018)	18/M	Falcine (frontal)	MMT	*EWSR1-CREM*
15	Konstantidinis et al. (2019)	13/F	Frontal	AFH	*EWSR1-ATF1*
16	Konstantidinis et al. (2019)	12/F	Frontal	AFH	*EWSR1-CREM*
17	Ghanbari et al. (2019)	58/F	Extra-axial (parietal)	AFH	*EWSR1-CREB1*
18	White et al. (2019)	9/M	Falcine (frontal)	MMT	*EWSR1-CREM*
19	Komastu et al. (2020)	Middle age/F	Intraventricular (3rd ventricle)	MMT	*EWSR1-CREB1*
20	Ballester et al. (2020)	67/M	Abutting dura (temporal)	MMT	*EWSR1-ATF1*

Conclusions

1. AFH shows a non-specific immunophenotype, but demonstrates recurrent *EWSR1-CREB* gene rearrangements
2. Intracranial AFH frequently shows myxoid change and shares histologic and genetic features with intracranial myxoid mesenchymal tumor
3. Rhabdoid features in AFH are rare and it is important to distinguish it from other mimics, particularly rhabdoid meningioma

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DLMP Mayo Clinic Rochester, MN

Howard Chang, M.D.

Dept. of Pathology, Sparrow Hospital, Lansing, MI


Cozumel, Mexico