AANP 2020
Diagnostic Slide Session
Case 1

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Vancouver CoastalHealth

64-year-old female:

1-year history of:

progressive right-sided dysesthesias beginning at biceps region

gait difficulties

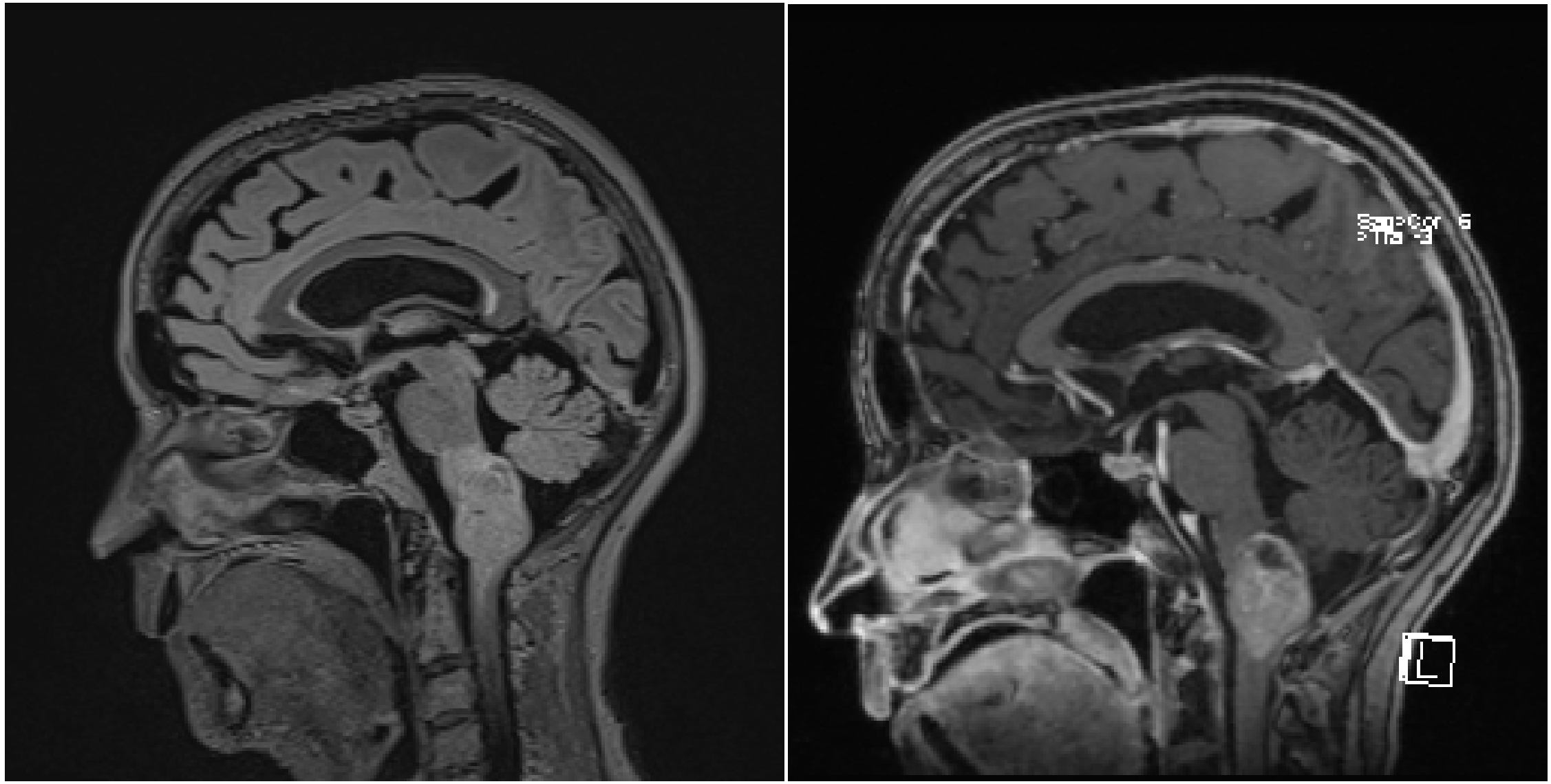
4-month history of :

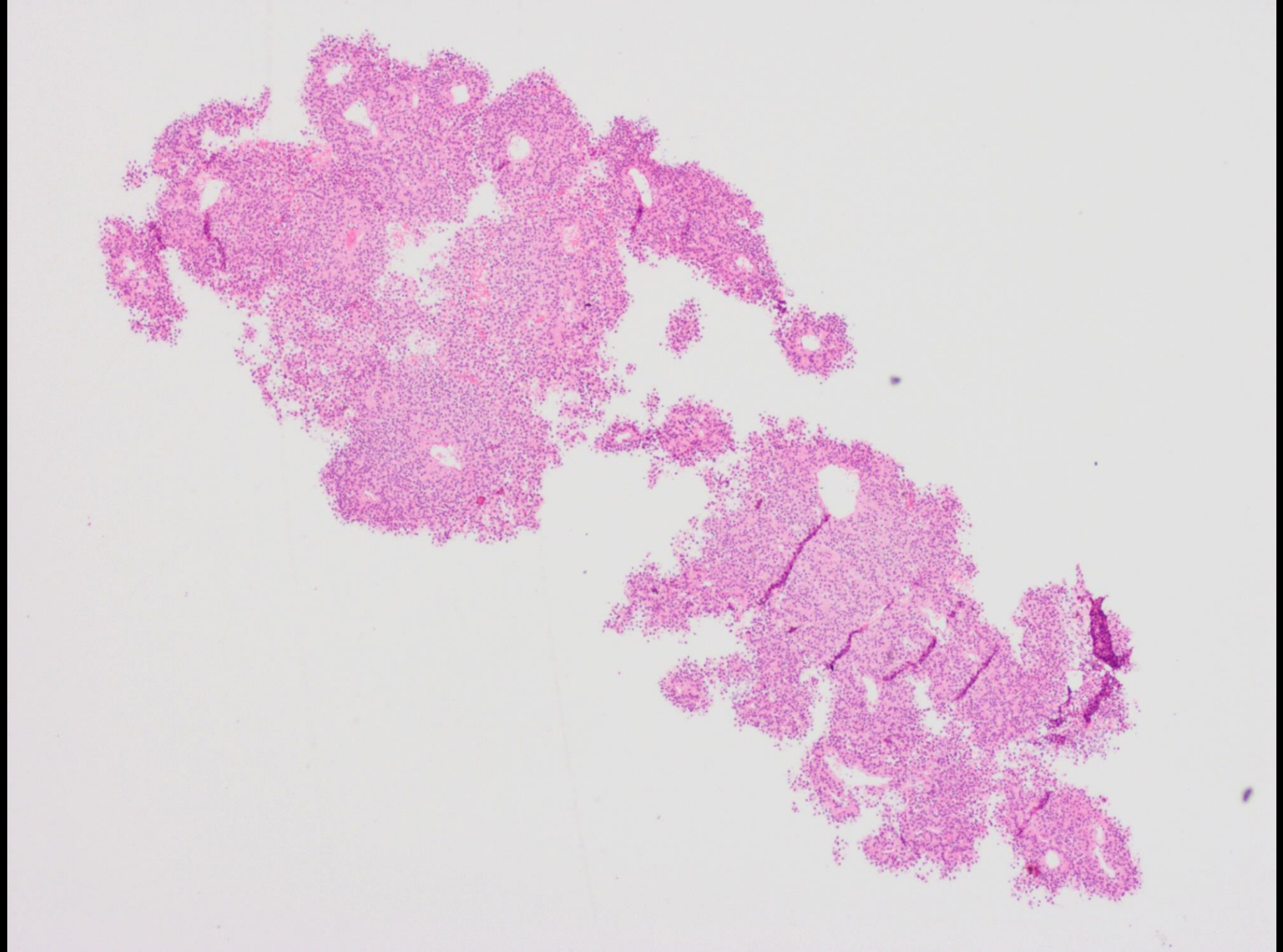
dysesthesias over entire right hemi-body sparing the face

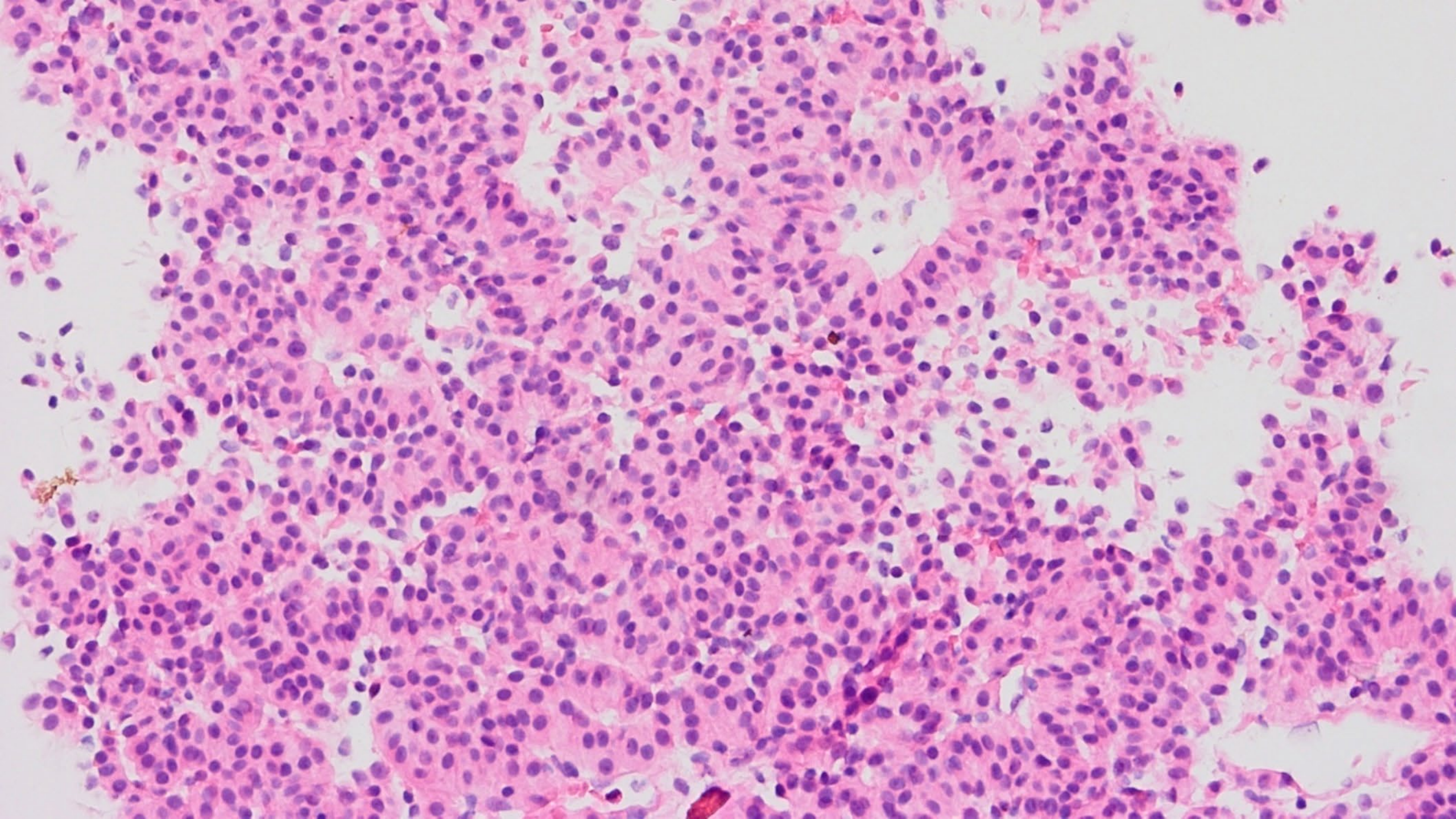
3-month history of:

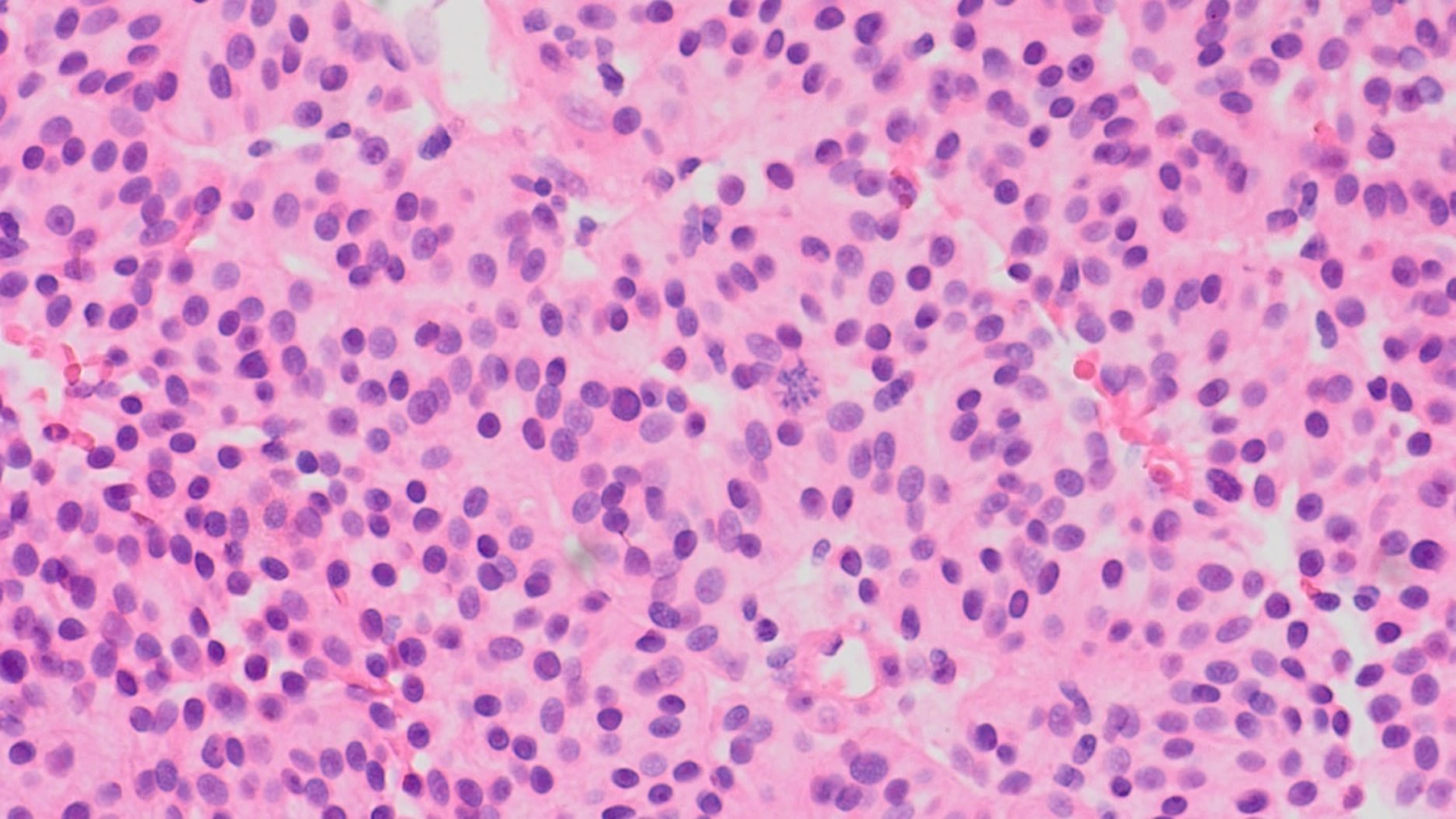
dysesthesia involves distal left lower limb

dysphagia





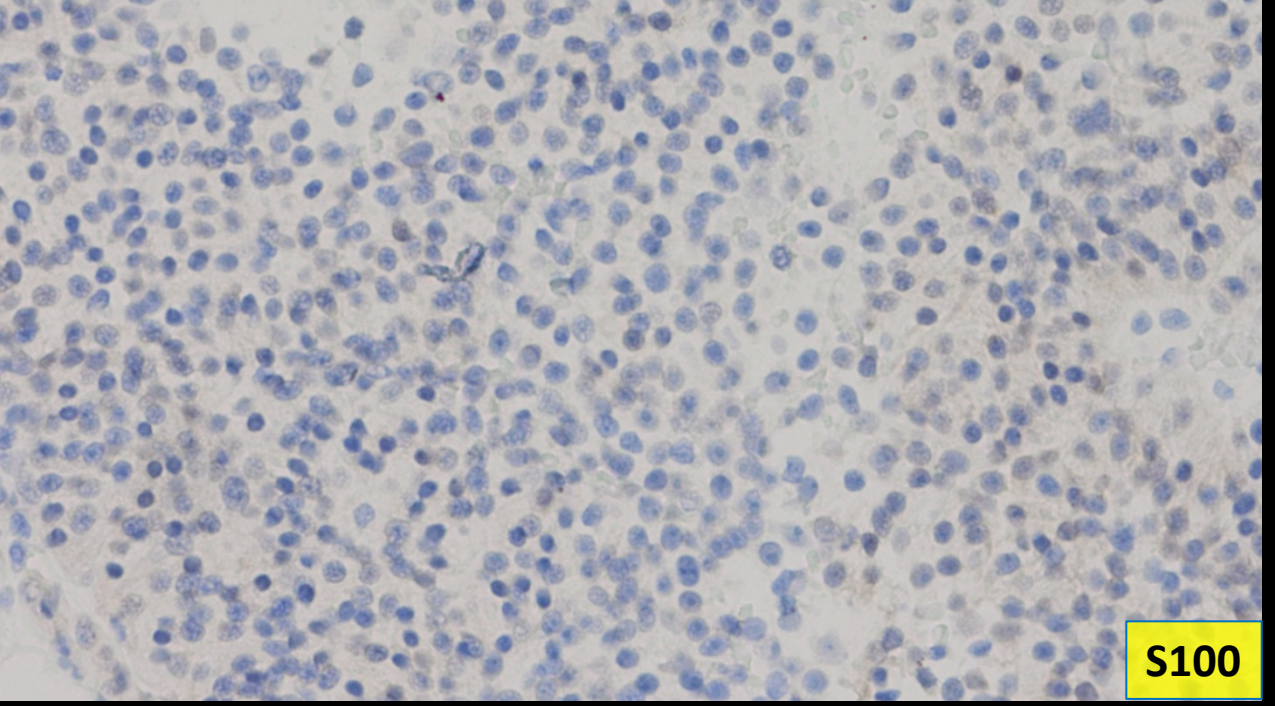


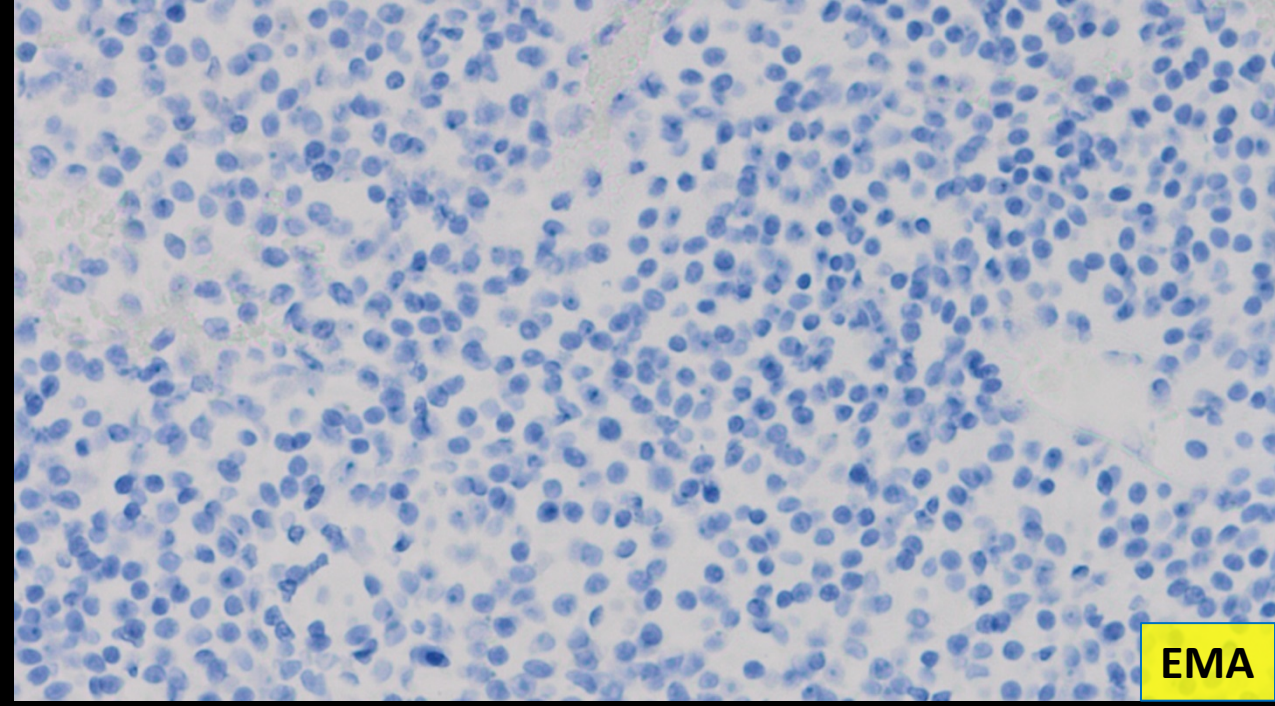


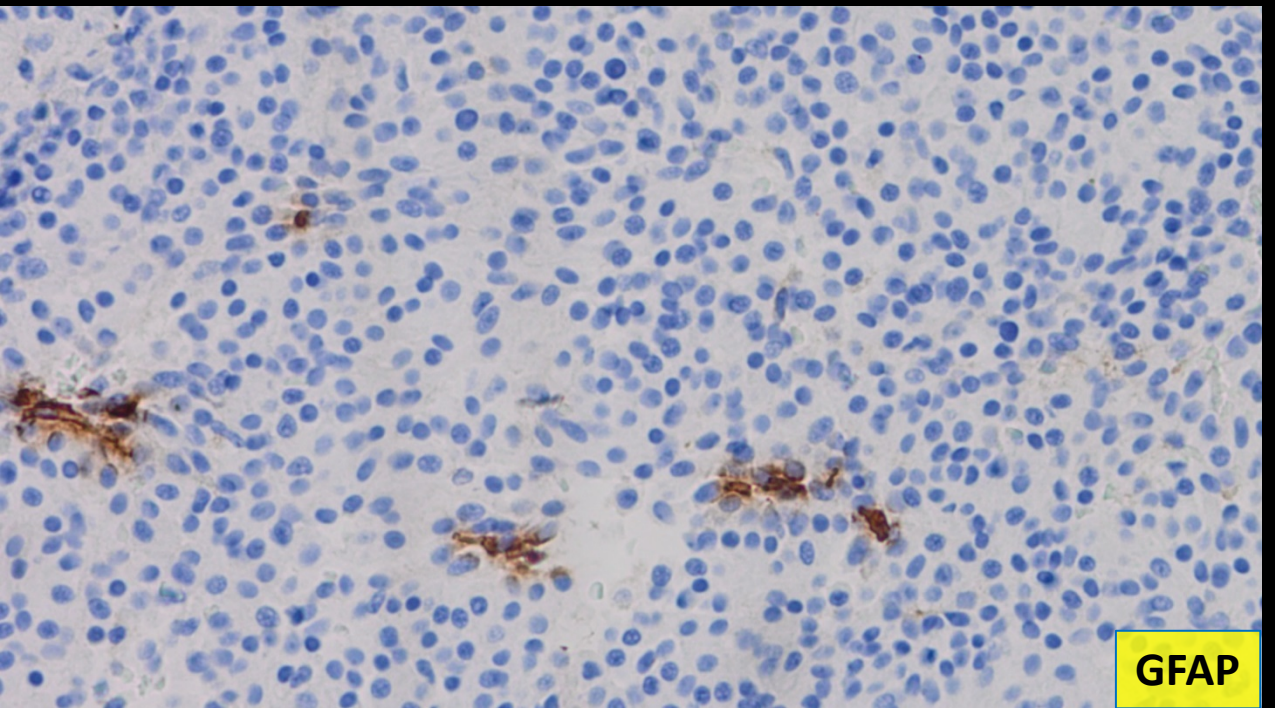
Differential?
Additional studies?

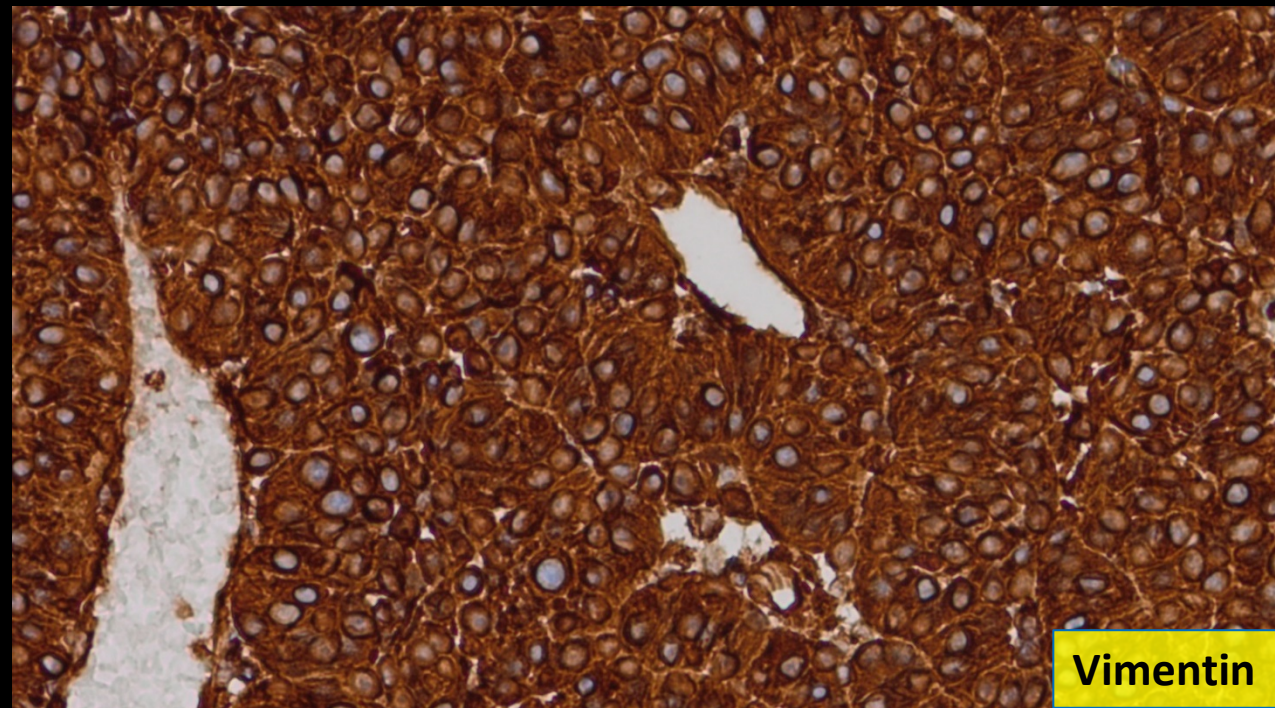
Differential – initial considerations

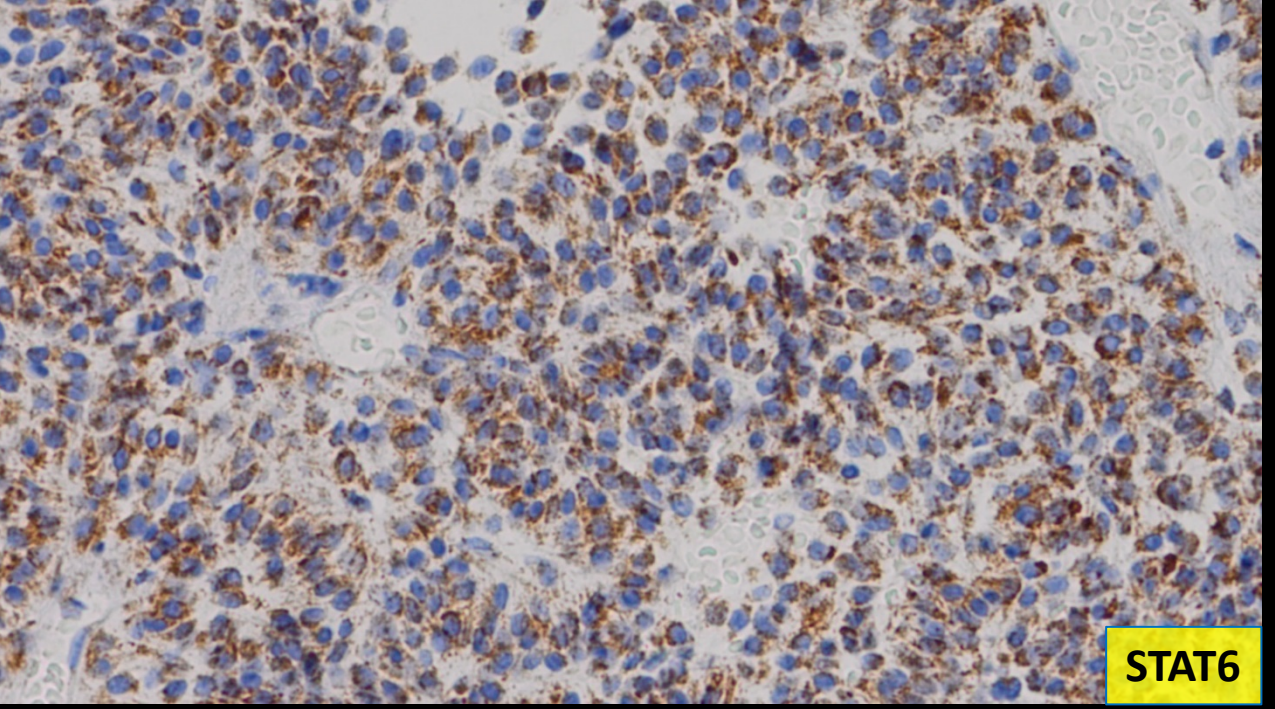
- Ependymoma
- Metastatic carcinoma
- Meningioma
- Hemangiopericytoma

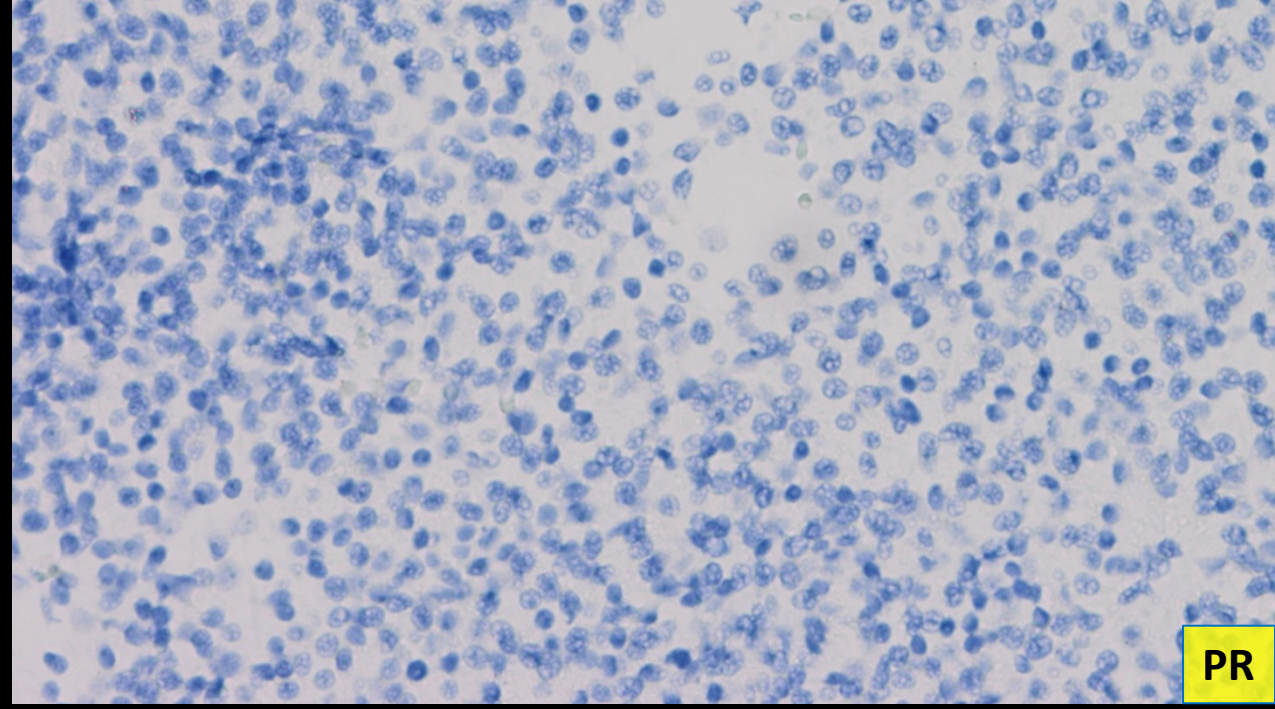

S100

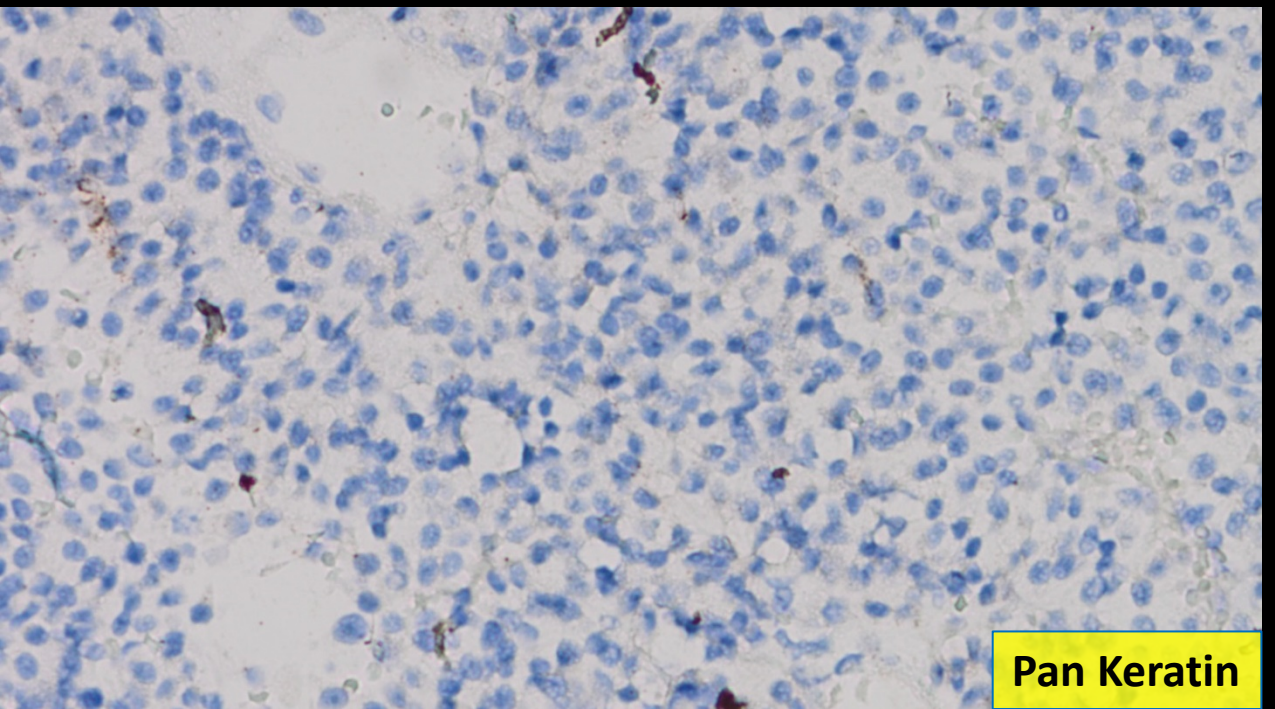

EMA

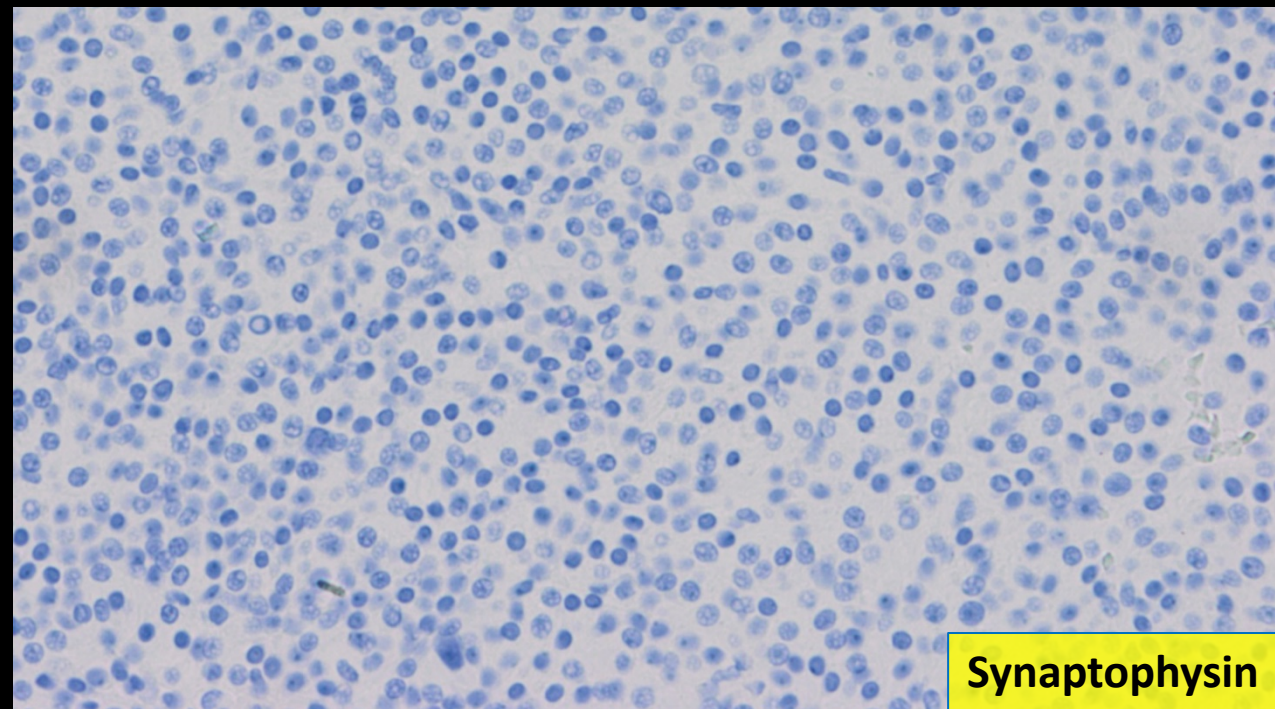

GFAP

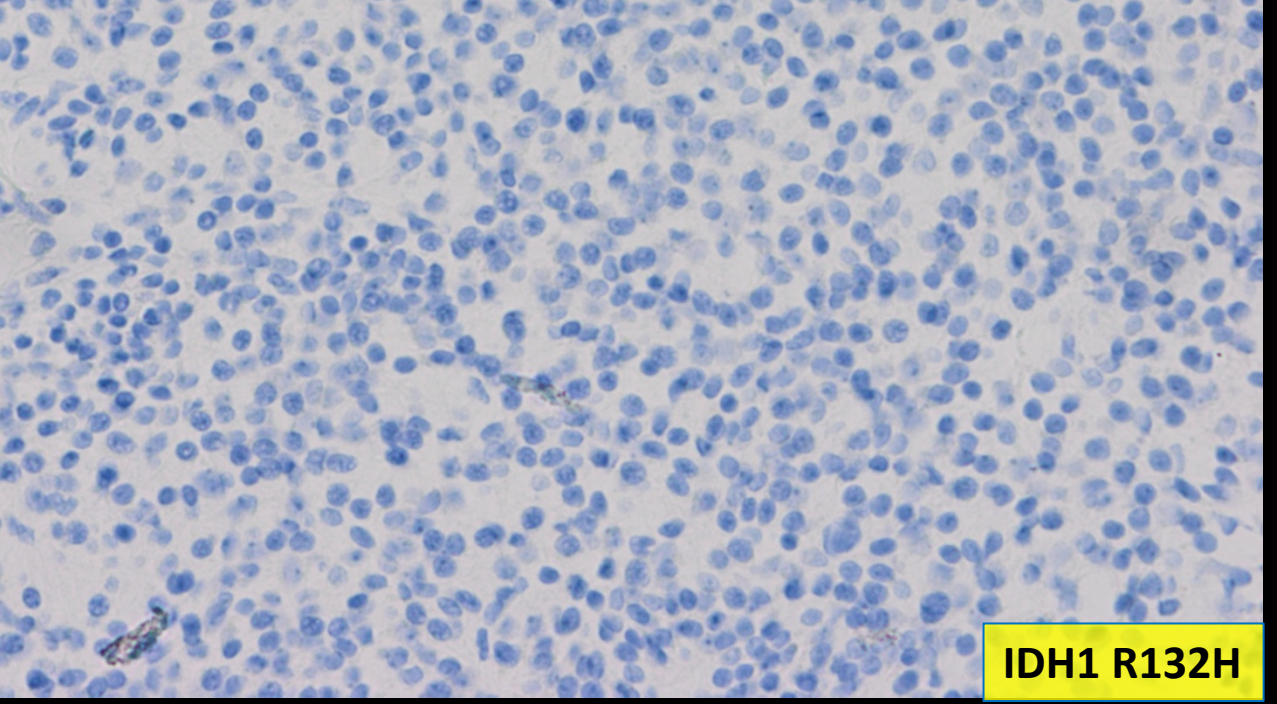

Vimentin


STAT6


PR


Pan Keratin


Synaptophysin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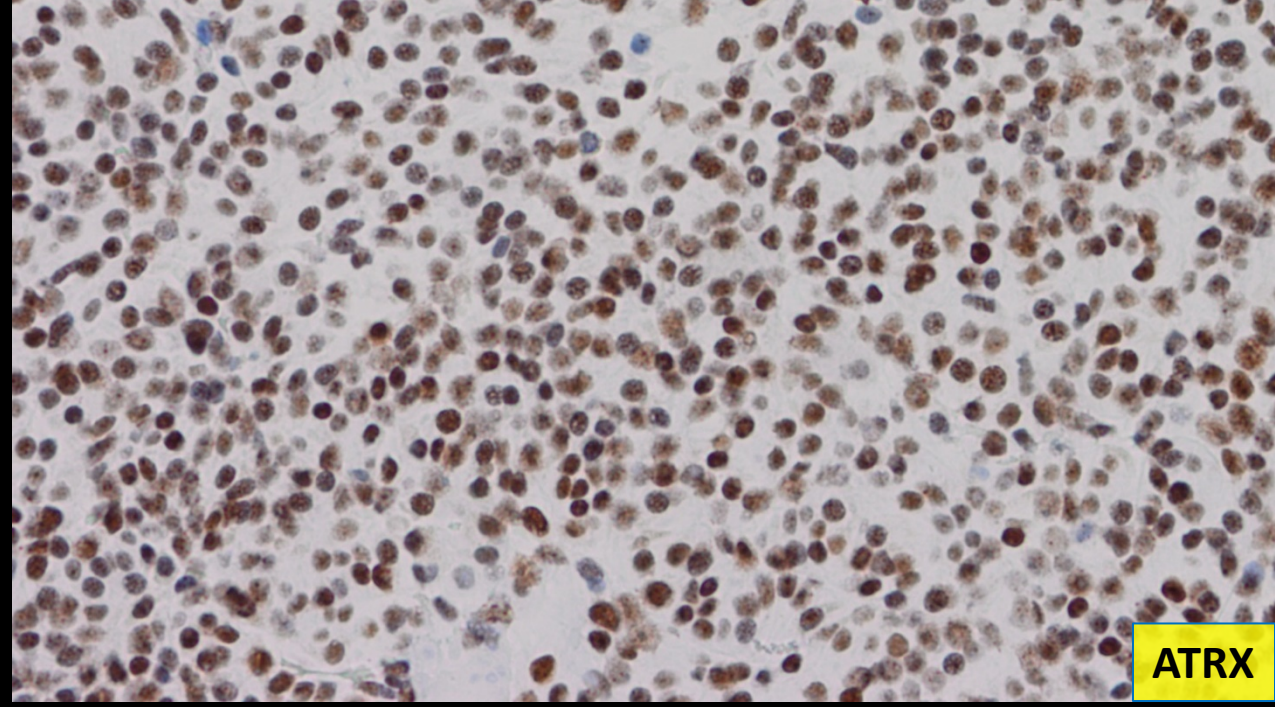
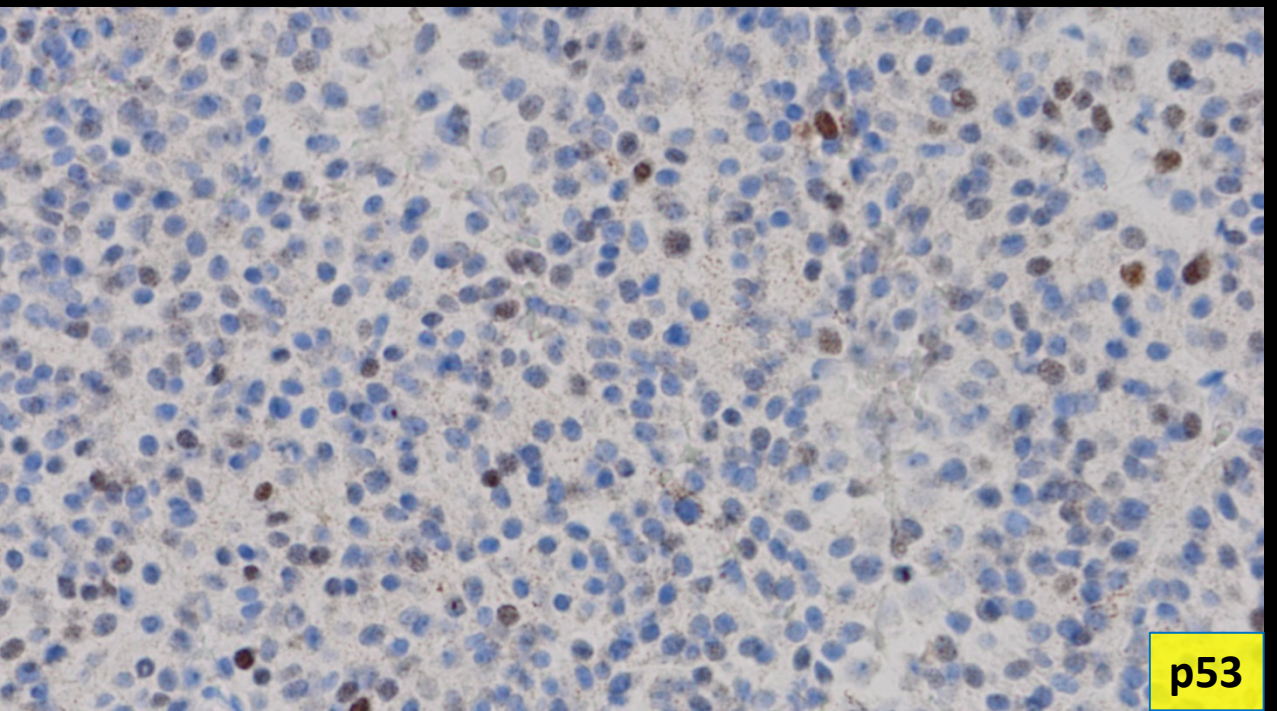
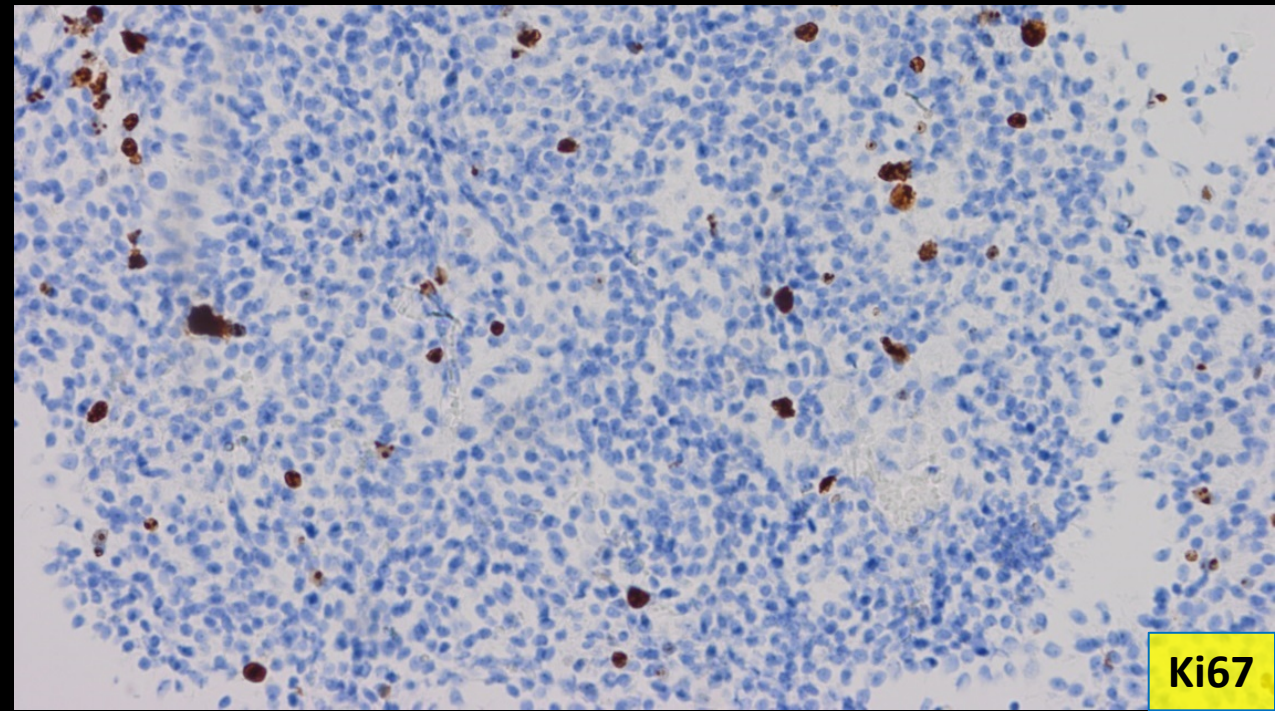
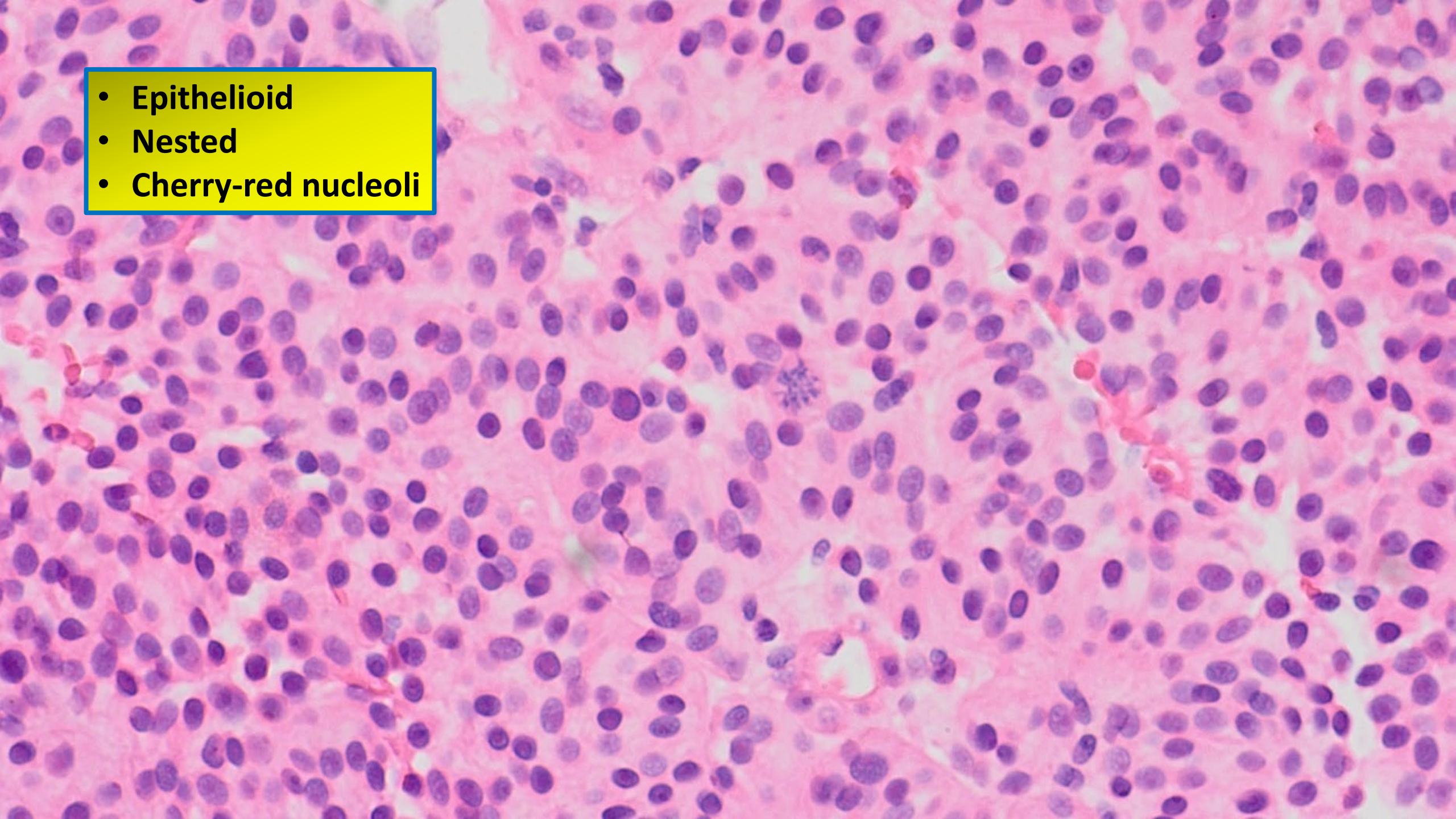
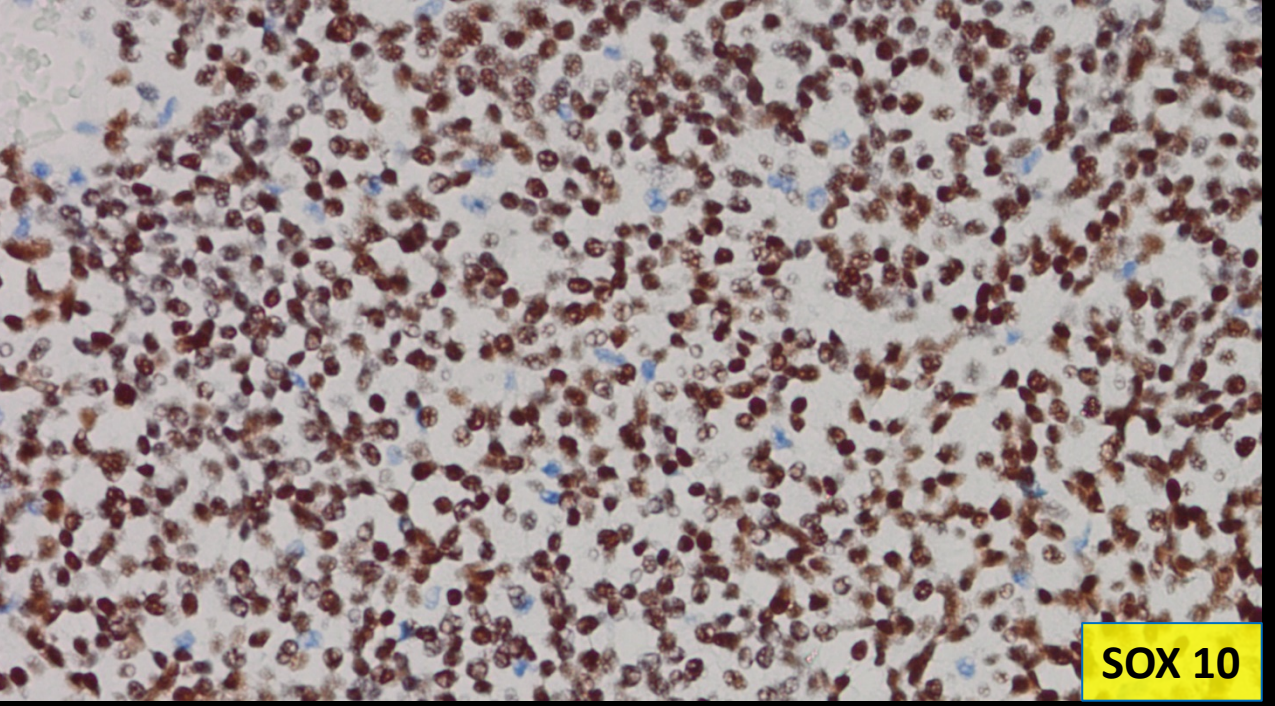
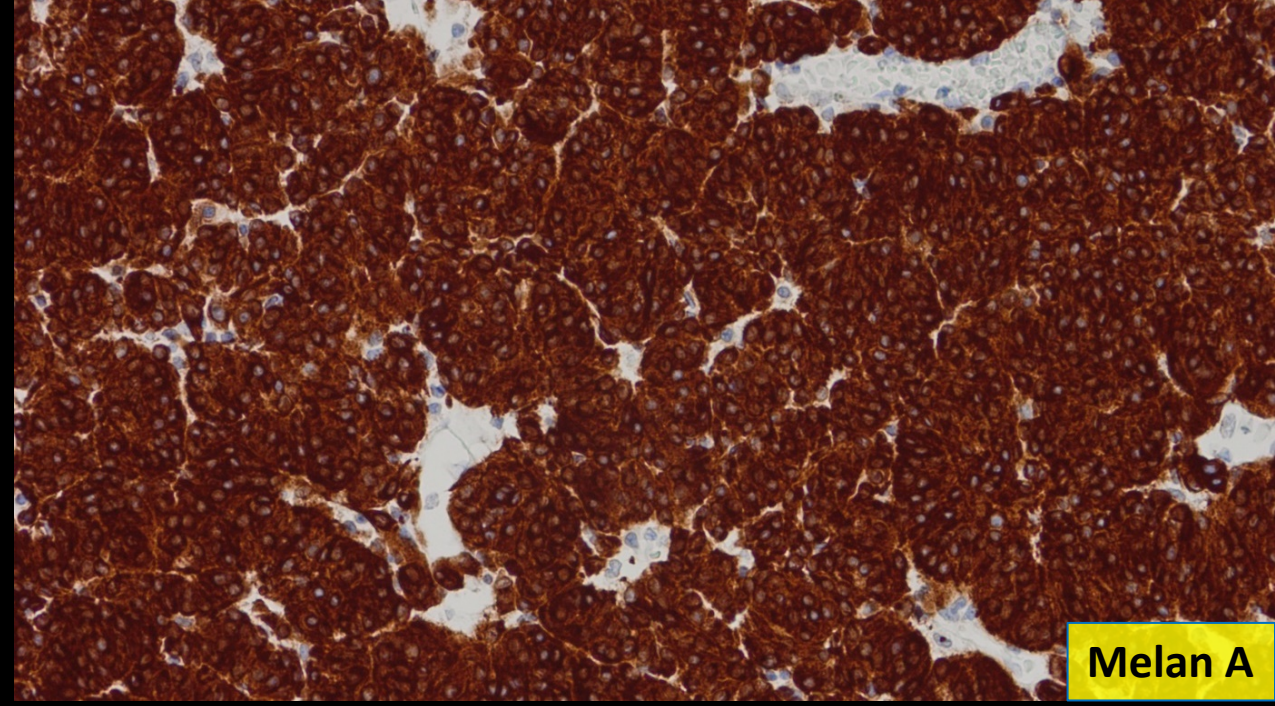

ATRX

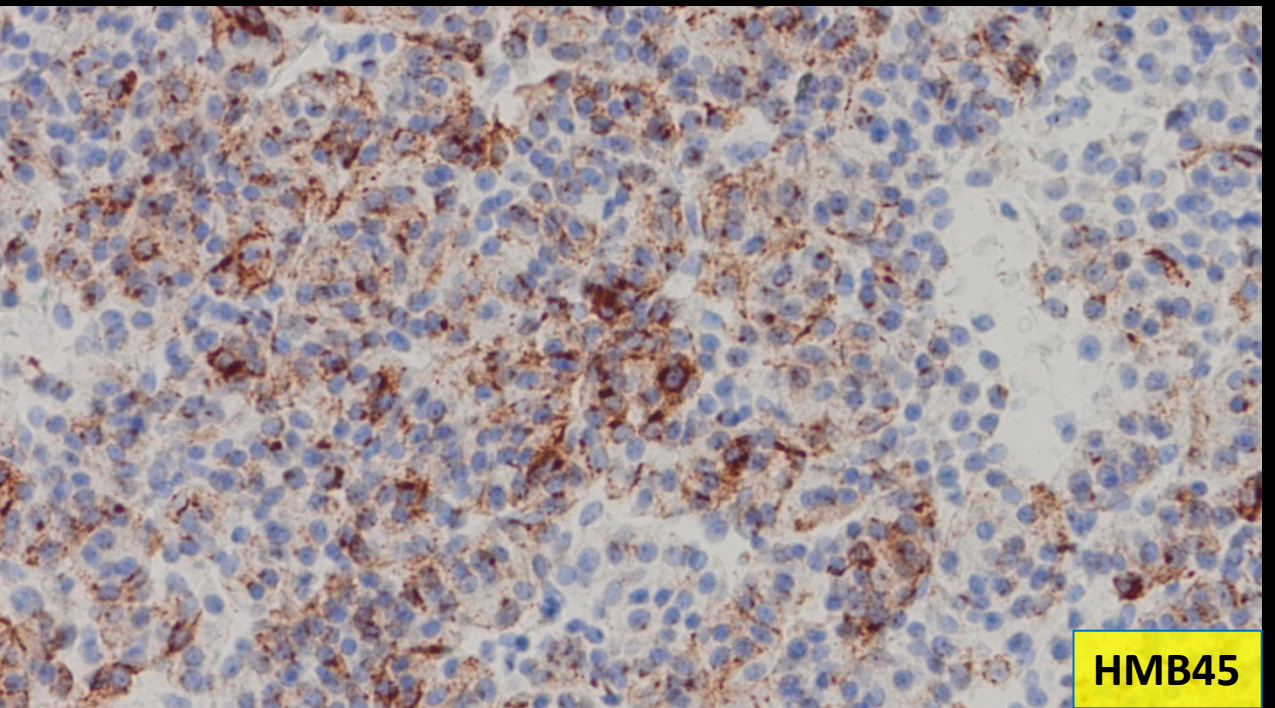

p53

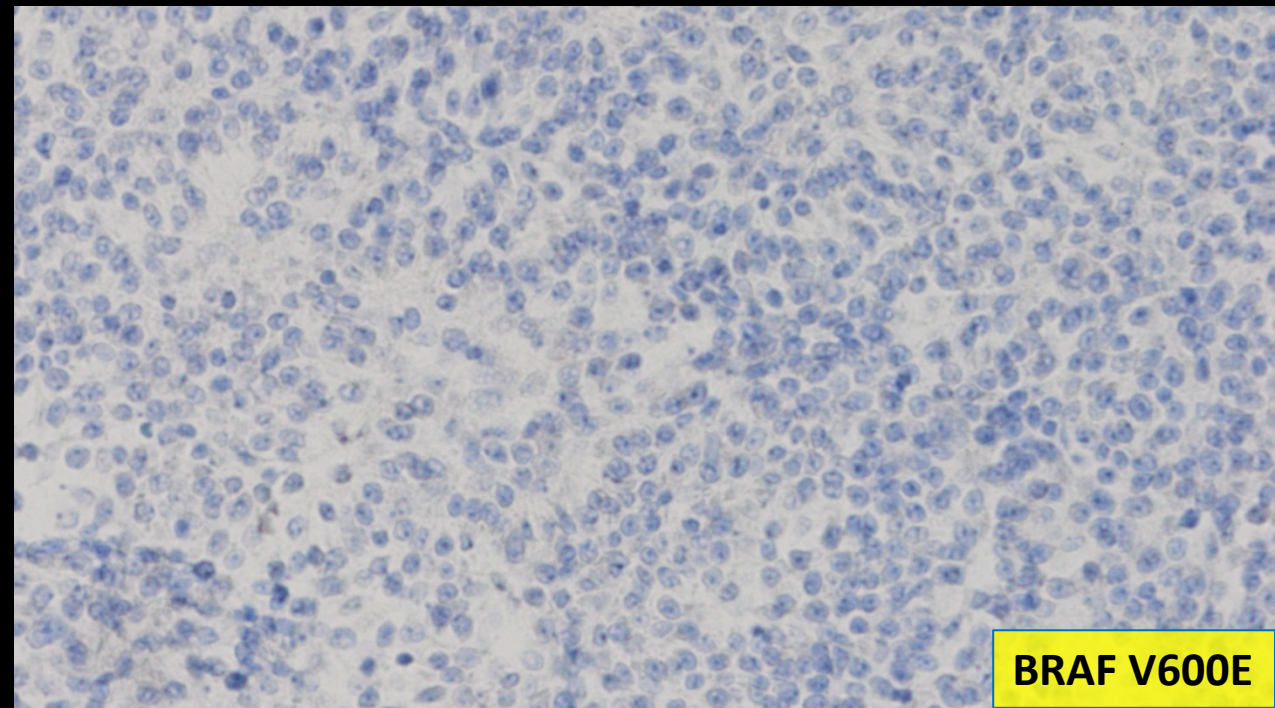

Ki67


• Epithelioid
• Nested
• Cherry-red nucleoli


SOX 10


Melan A


HMB45


BRAF V600E

Summary of Immunohistochemistry

Immunopositive

- Vimentin
- SOX10
- Melan A
- HMB45

Ki67: 2-5%

ATRX (retained)

P53 (wildtype)

Immunonegative

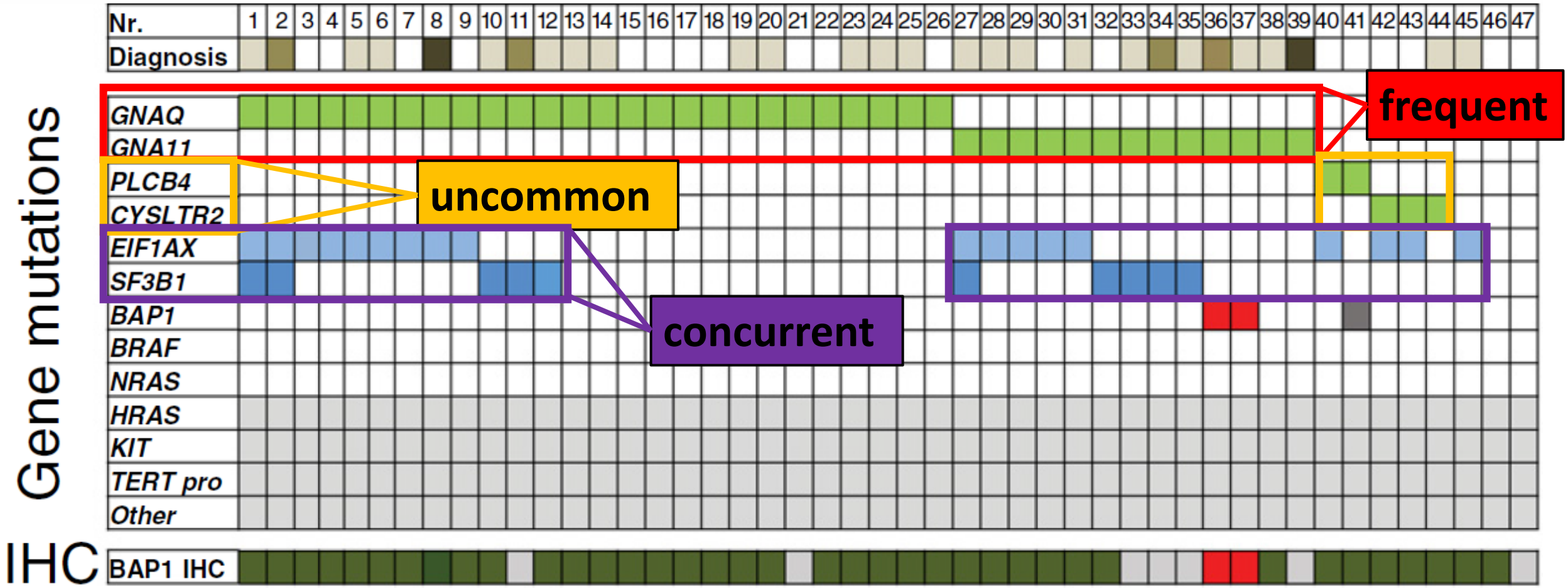
- BRAF V600E
- GFAP
- IDH1
- CD56
- EMA
- PR
- Synaptophysin
- CD99
- TTF1
- CAM 5.2
- CK
- PAX8
- Desmin
- SMA

Initial diagnosis:

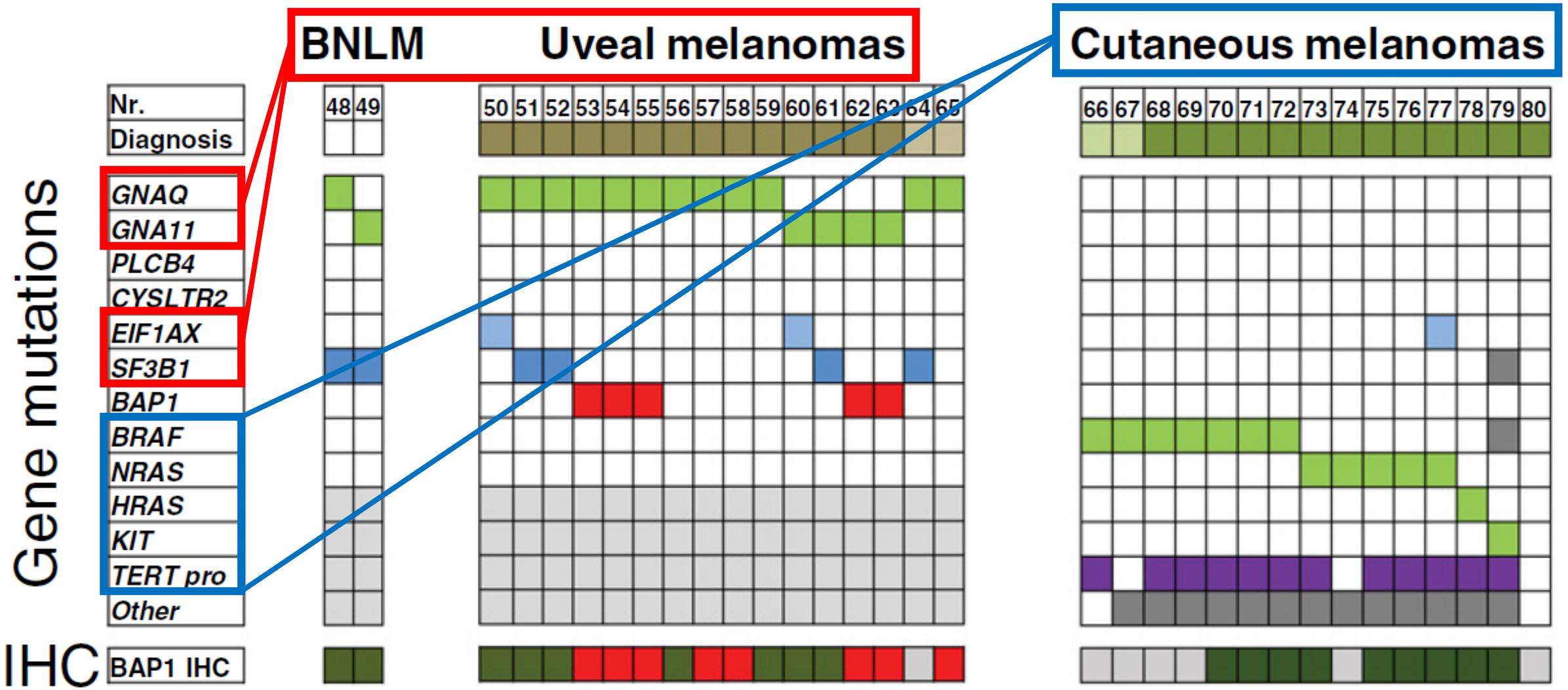
melanocytic neoplasm

metastatic vs primary?

Primary leptomeningeal melanocytic tumors



*Clin Cancer Res; 24(18) September 15, 2018



BNLM: blue nevus-like melanoma

*Clin Cancer Res; 24(18) September 15, 2018

Molecular genetics:

- Our case sent for next generation sequencing
- **GNA11:**
- Variant of functional significance detected.
- GNA NM-002067.4:c.626A>T.p.(Gln209Leu)(VAF:48.1%)

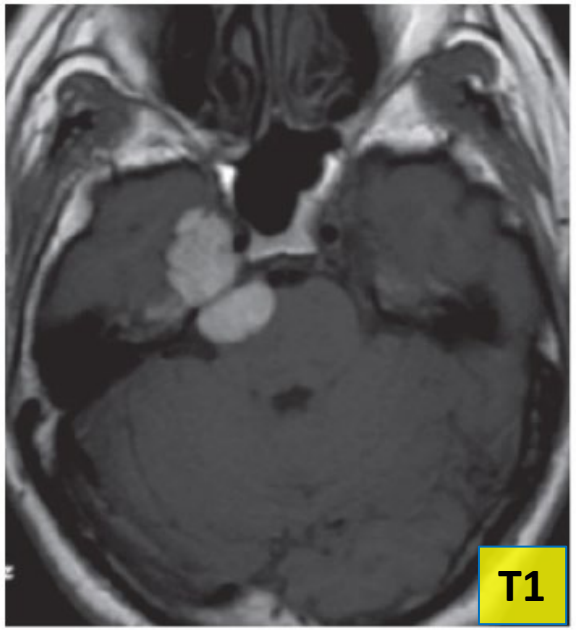
- No reportable variants were noted in **BRAF, KIT and GNAQ**

Diagnosis

Amelanotic primary meningeal melanocytoma

Primary melanocytic tumors

- Very rare – 0.06 to 0.1% of primary brain tumors
- Most common location – brainstem/spinal cord
- Detectable with IHC
  - HMB45, MelanA, S100, SOX10
- Electron microscopy:
  - melanosomes/premelanosomes
  - partial to no basement membrane
  - no desmosomes



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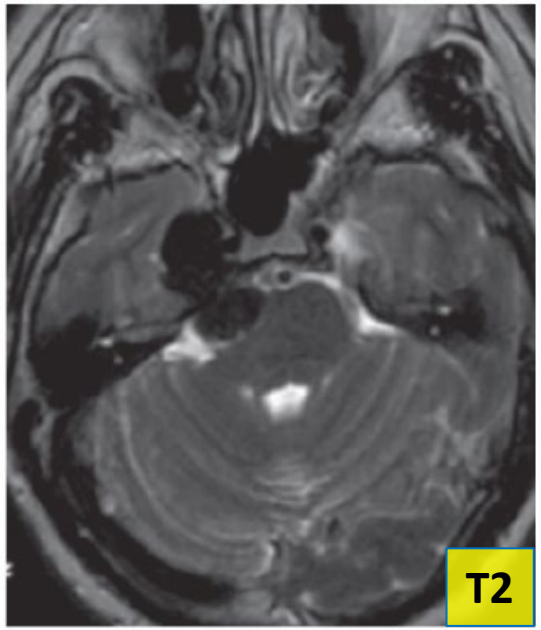

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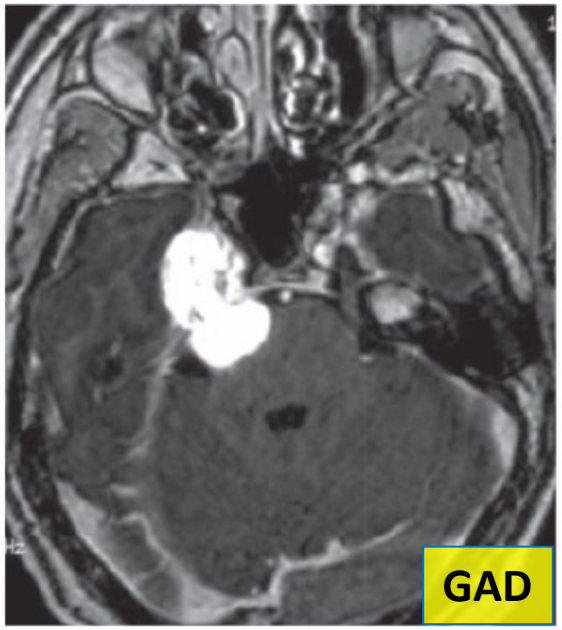
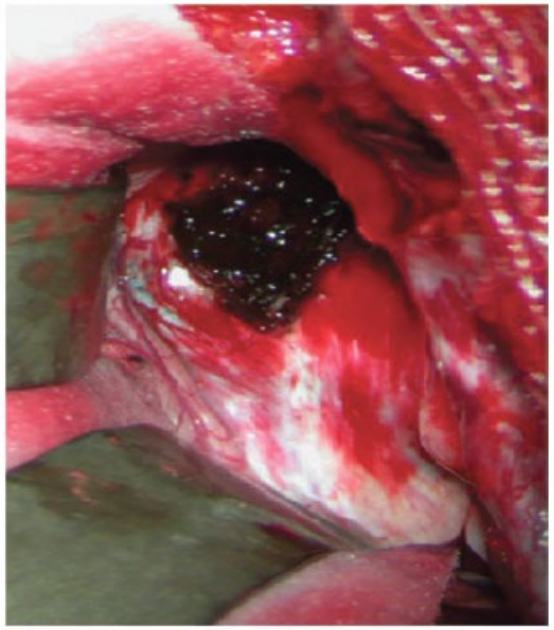
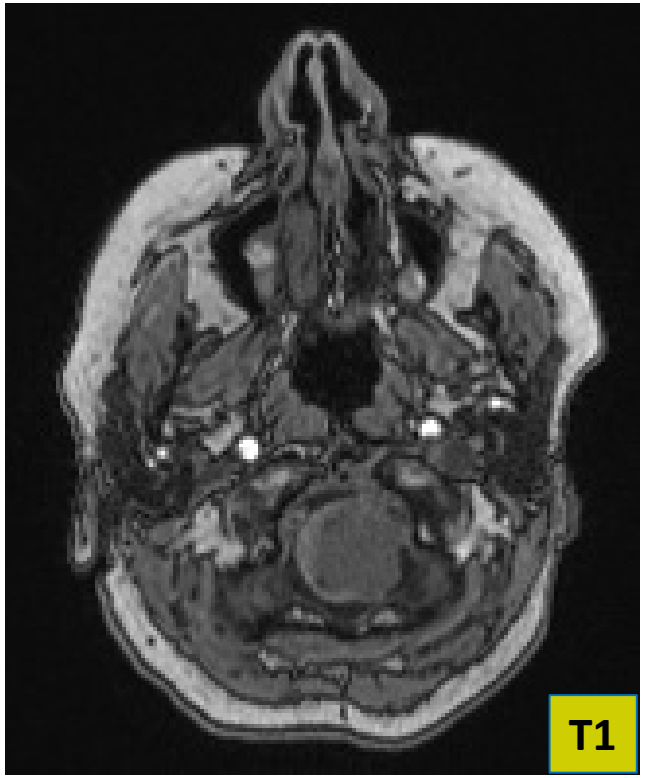


c.



d.

*Radiographics; 29(5):1503-24. Sep-Oct 2009



Follow up:

- Symptomatic improvement
- Immune checkpoint inhibitors
  - 3 of 4 cycles: ipilimumab and nivolumab
- Complications – now resolved
  - lymphocytic colitis
  - maculopapular rash
- No evidence of tumor progression

Summary: Amelanotic melanocytoma

- Very rare, significant diagnostic challenges
- MRI: useful for pigmented tumors
- Diagnosis: confirmed by immunohistochemistry
- Molecular genetics: GNA11 mutation confirmed primary CNS melanocytoma diagnosis

References

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3. Lach B, Reddy K, Sommer DD, et al. Amelanotic melanocytoma of the sella mimicking pituitary adenoma. Clin Neuropathol. 2017 Nov/Dec;36(6):283-287.
4. Jun Ma, Zhong Zhang, Shu Li, et al. Intracranial amelanotic melanoma: a case report with literature review. World J Surg Oncol. 2015;13:182.
5. Smith AB, Rushing EJ, Smirniotopoulos JG. Pigmented lesions of the central nervous system: radiologic-pathologic correlation. Radiographics. 2009 Sep-Oct;29(5):1503-24.


Thank you!
Questions?