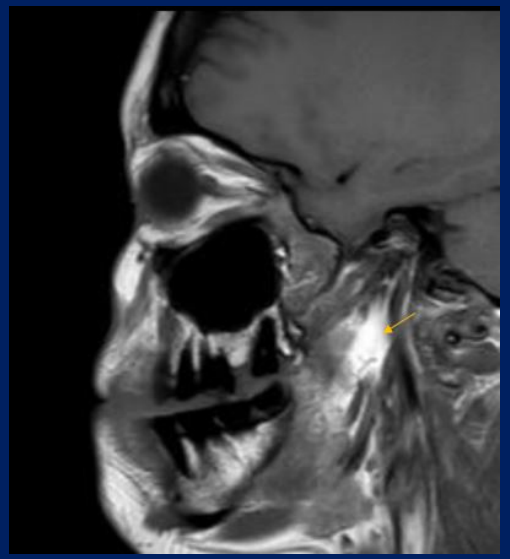
Case 2019-9

J. Stephen Nix MD, Lisa M. Rooper MD, Analiz Rodriguez MD, PhD,
Murat Gokden MD

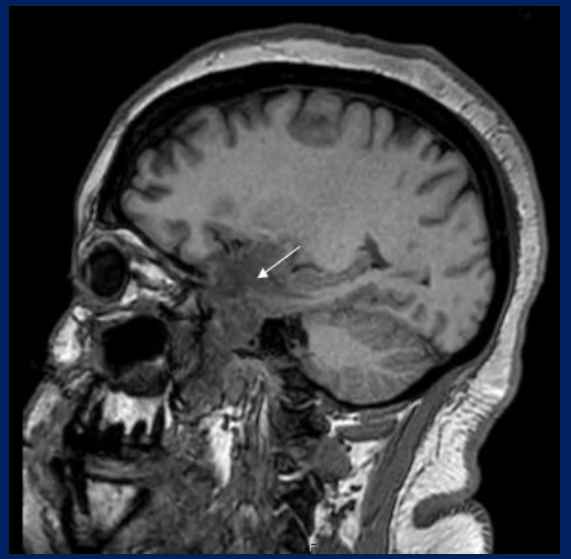
No disclosures

Clinical History and Imaging

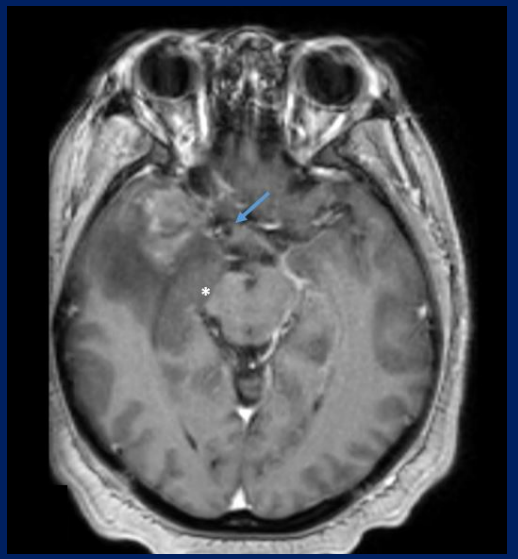
- Woman in 40s presents with right-sided headache, heaviness of the head, tongue tingling, numbness, and vomiting
- Imaging reveals mass involving V2 division of trigeminal nerve, eroding into the right maxillary sinus, and expanding into the right foramen rotundum with vasogenic edema of the temporal lobe



Left panel sagittal T1 hyperintense lesion. Initial presentation.

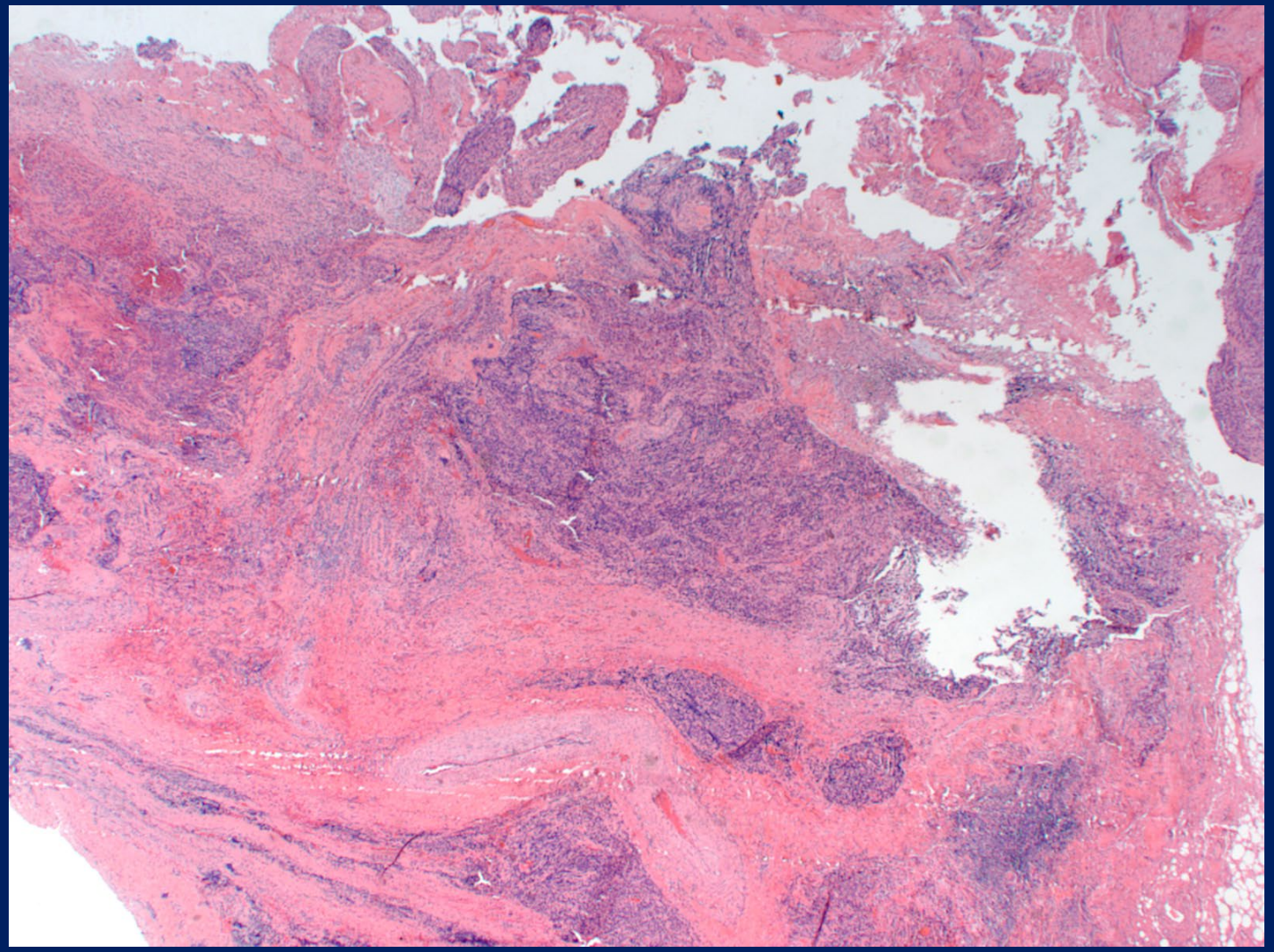


Recurrence with brain involvement. Sagittal T1



Axial post-contrast T1 before 2nd surgery. R temporal lobe involvement (arrow) and peripheral edema (*).

Initial Resection



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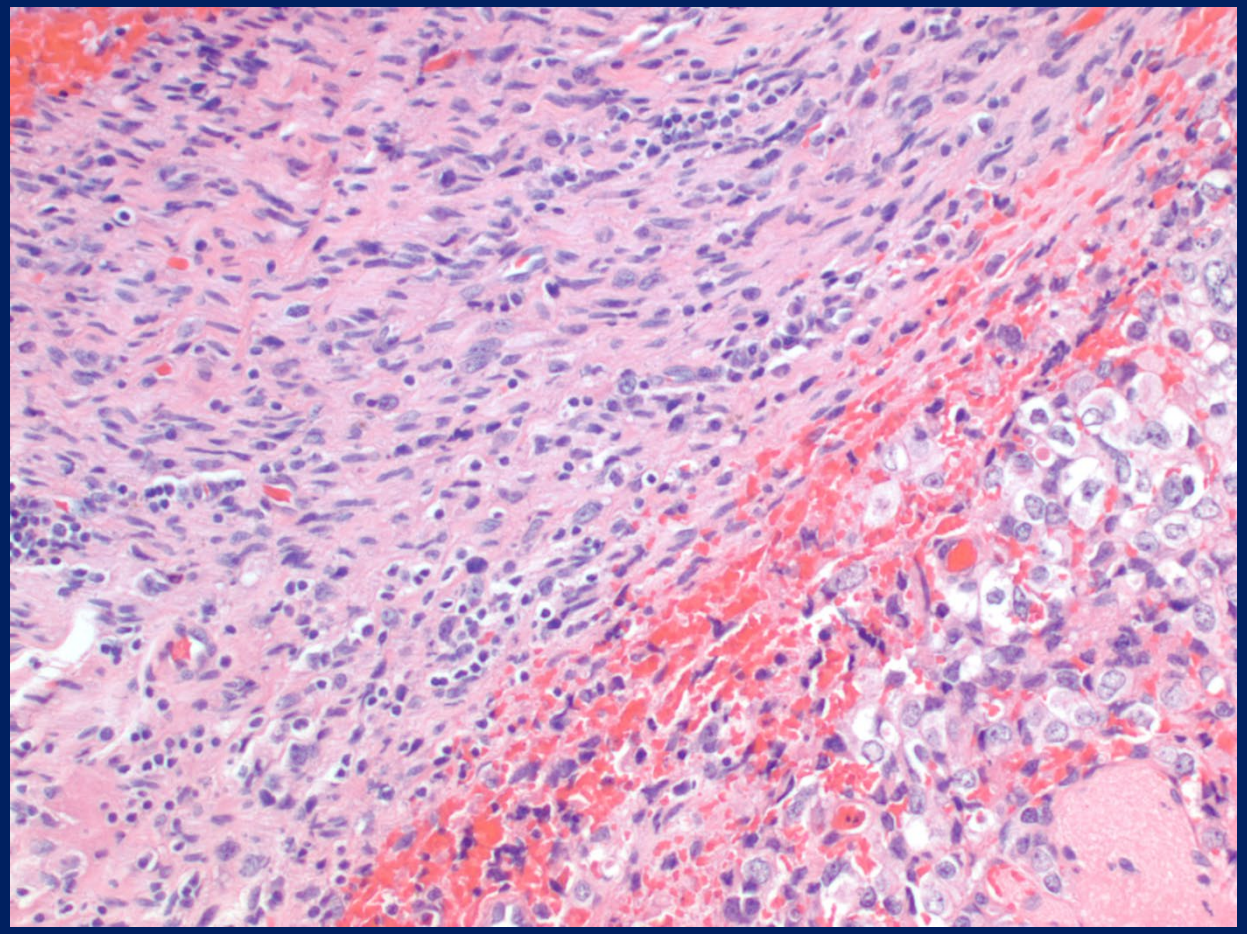
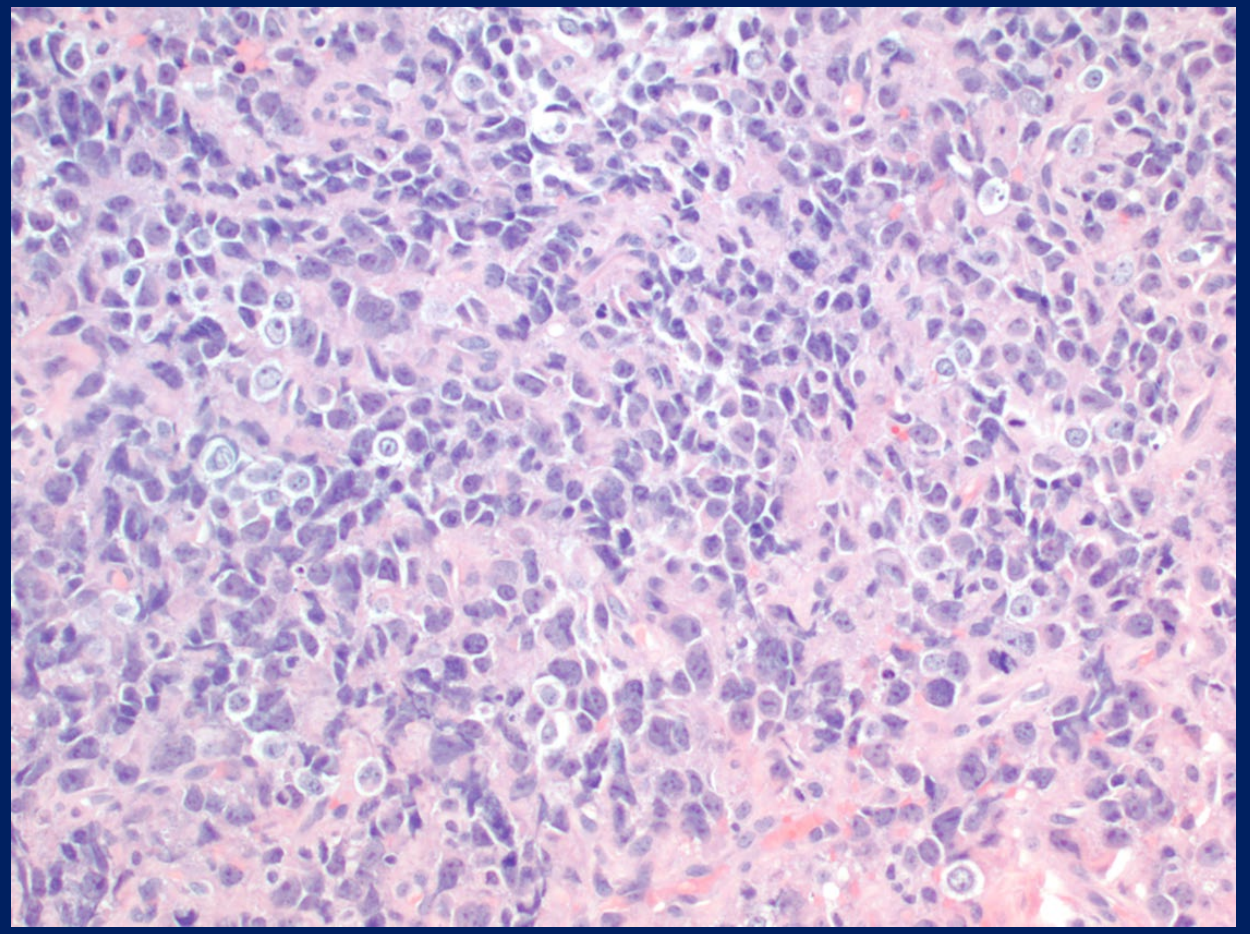
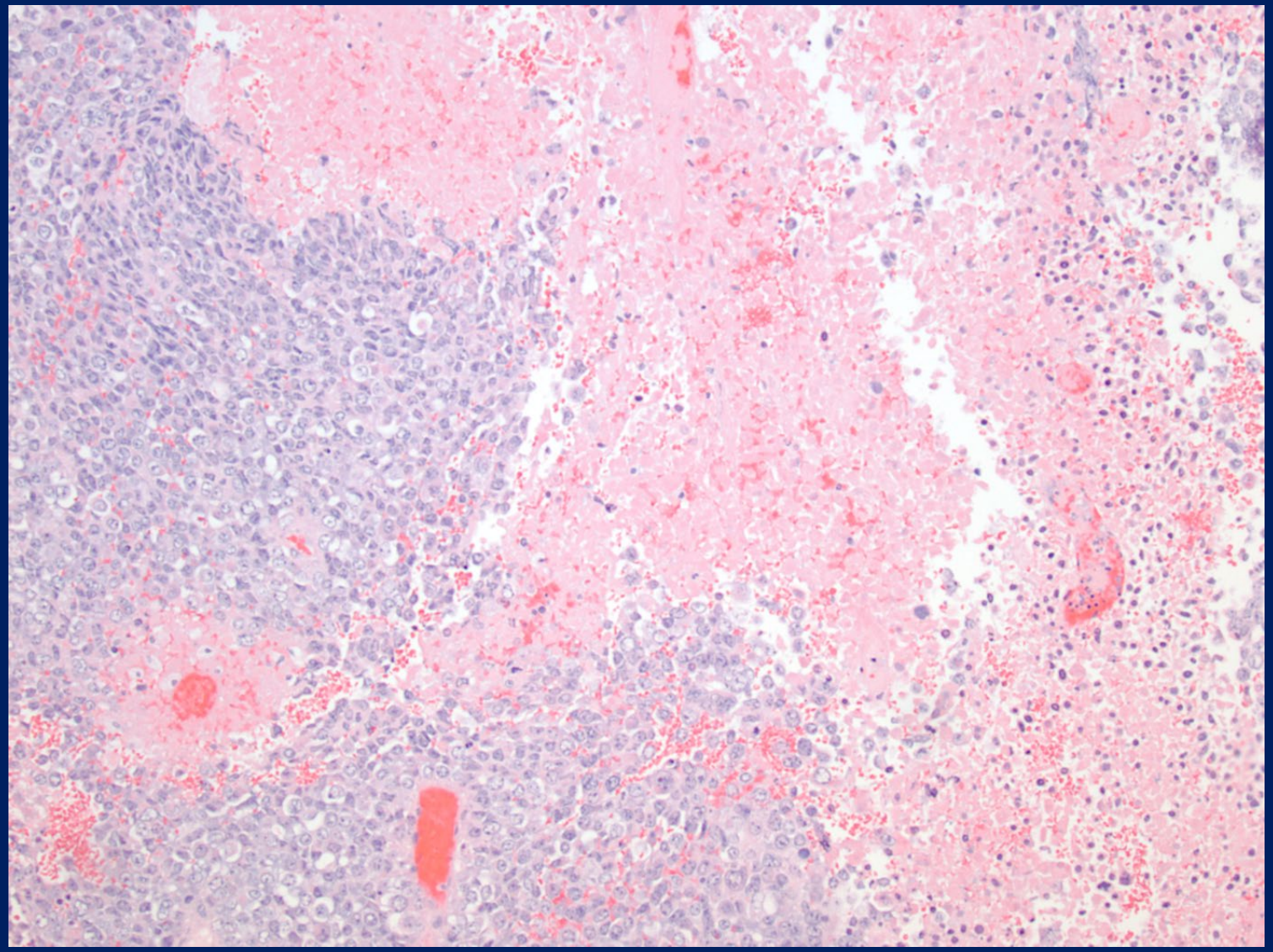
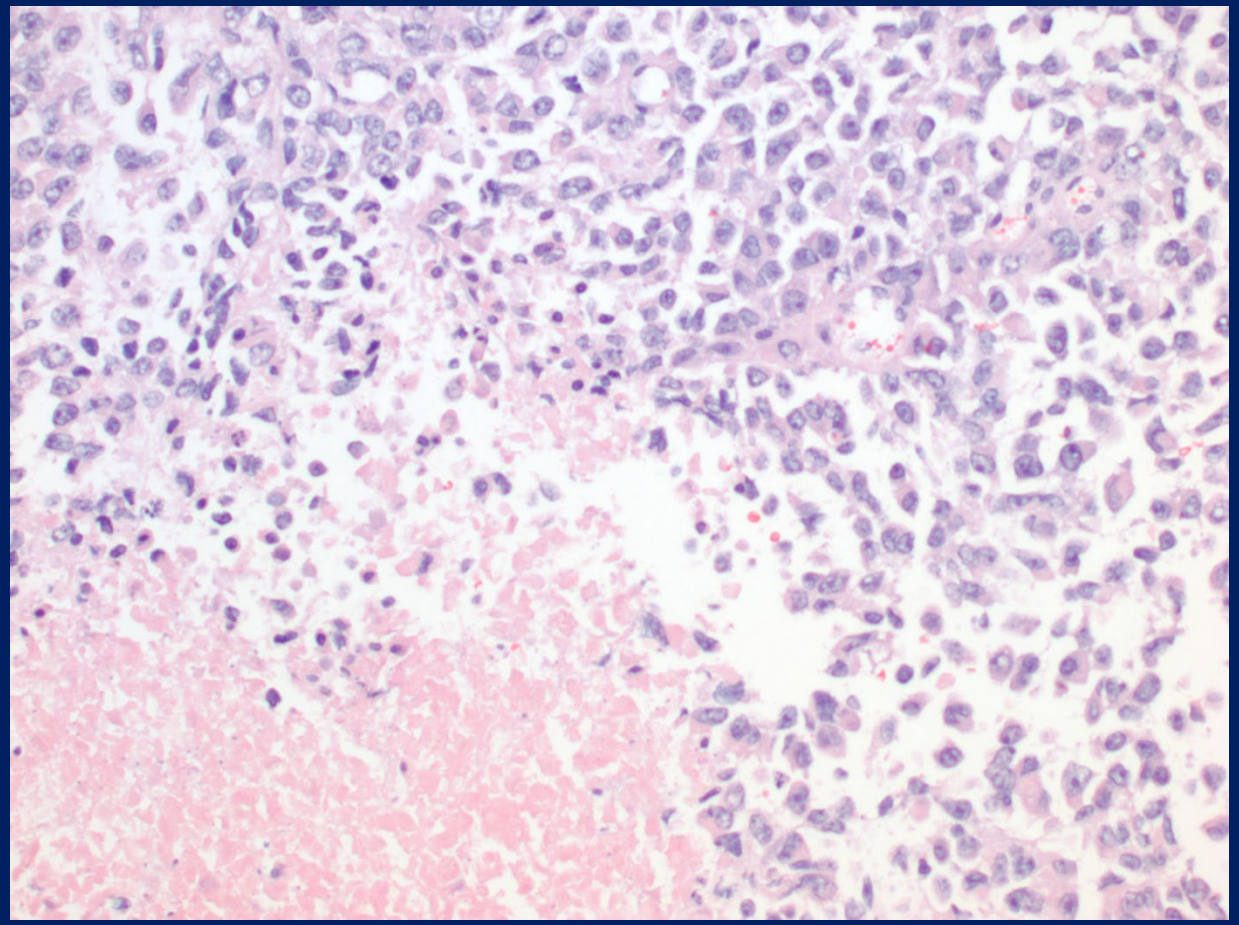


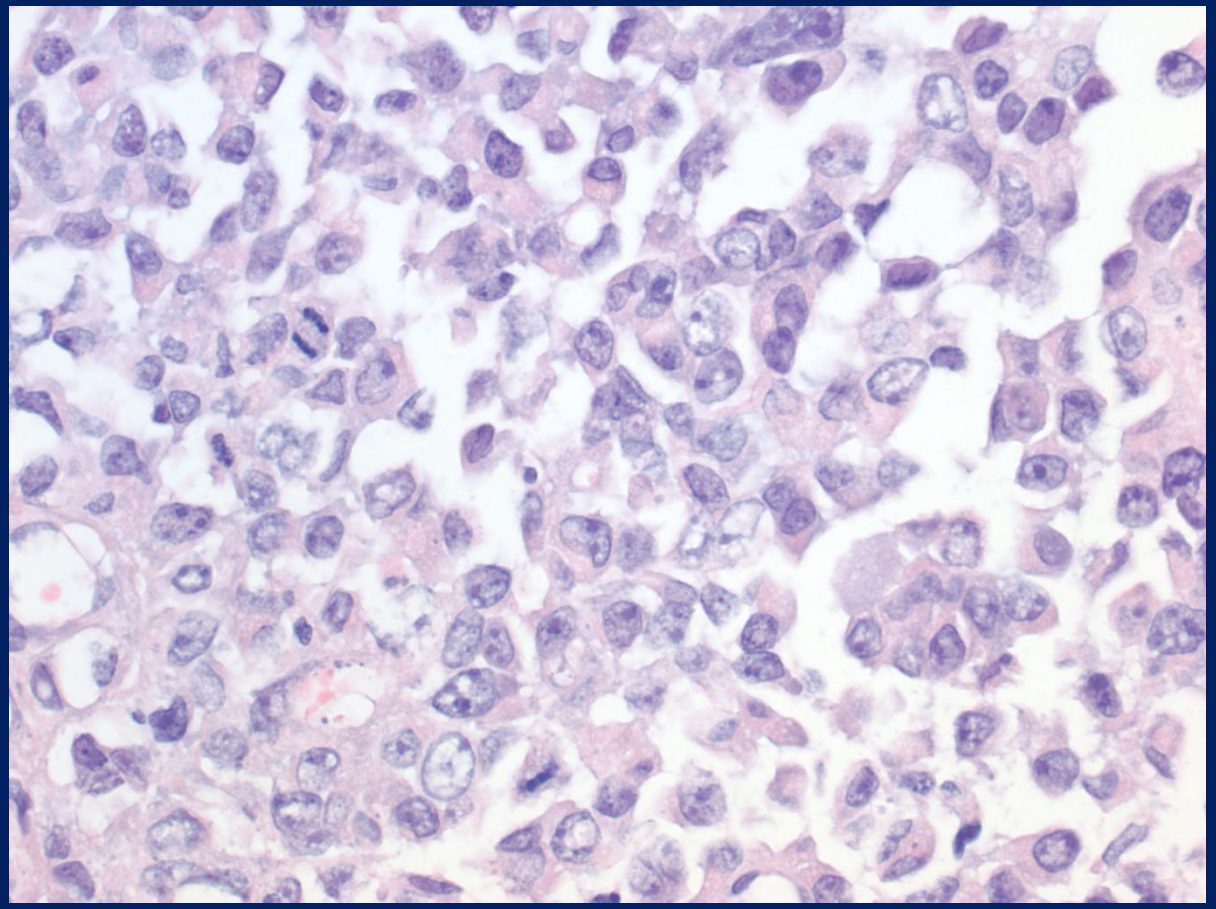


Second Resection



Second Resection

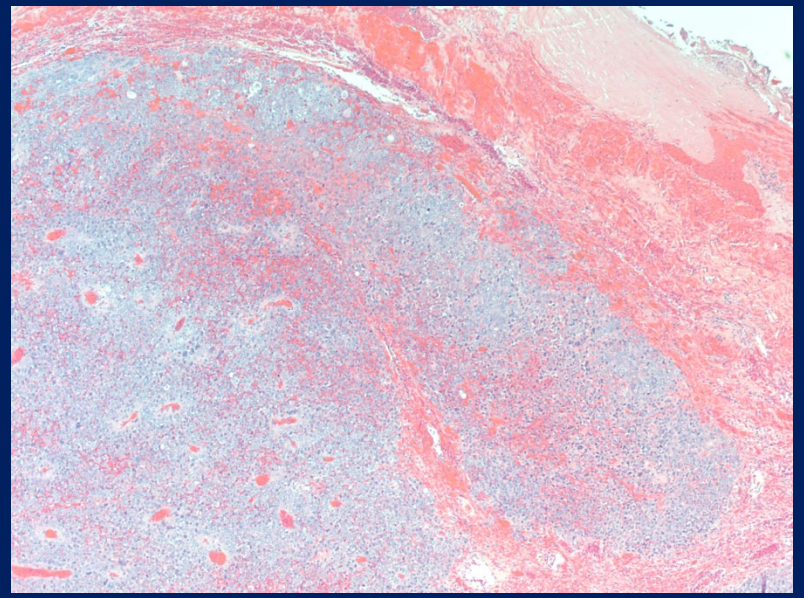
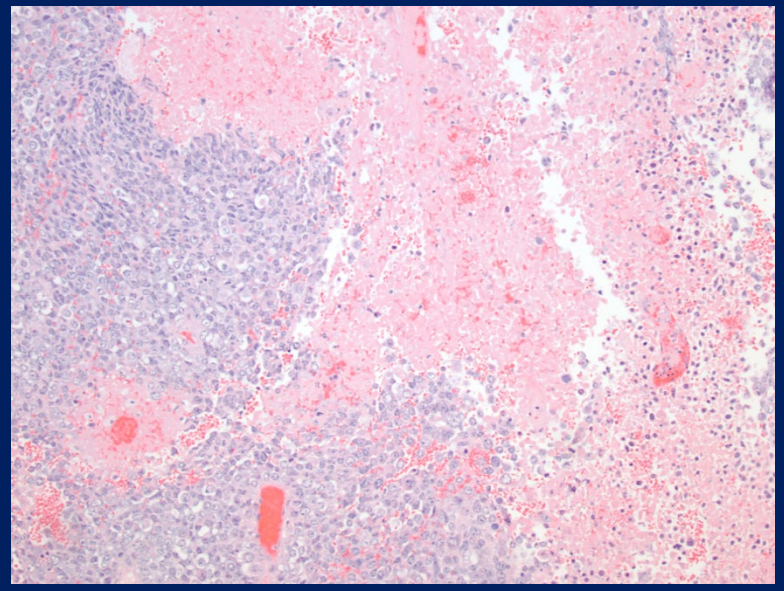
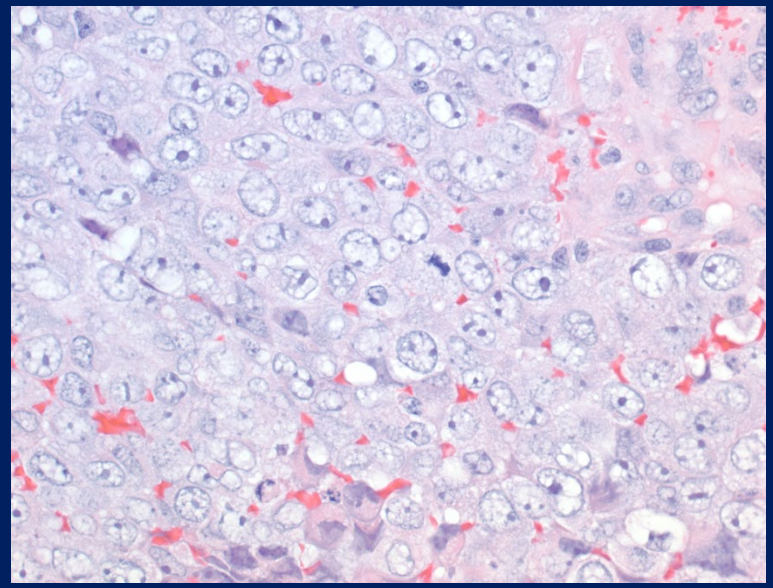
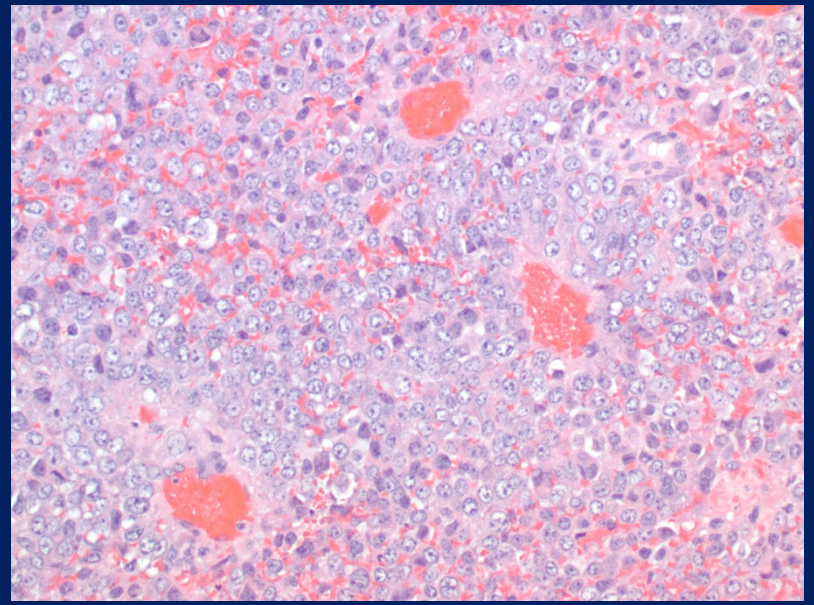
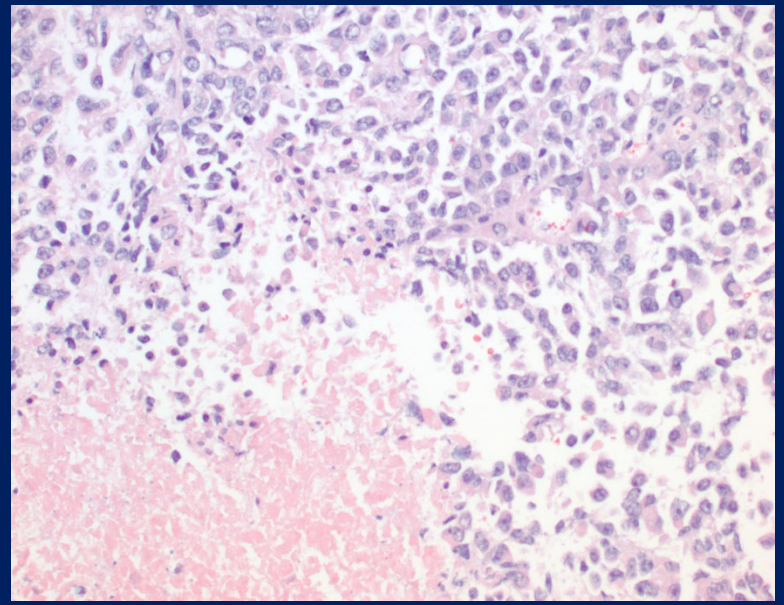
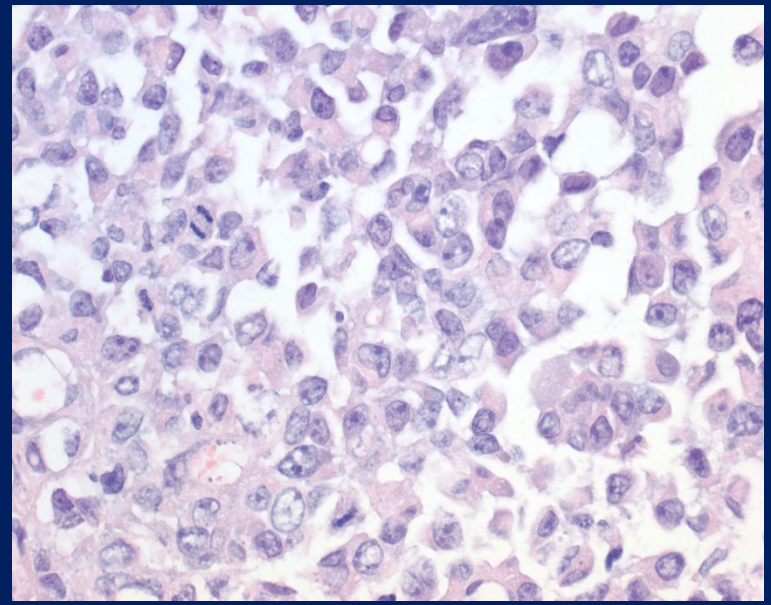


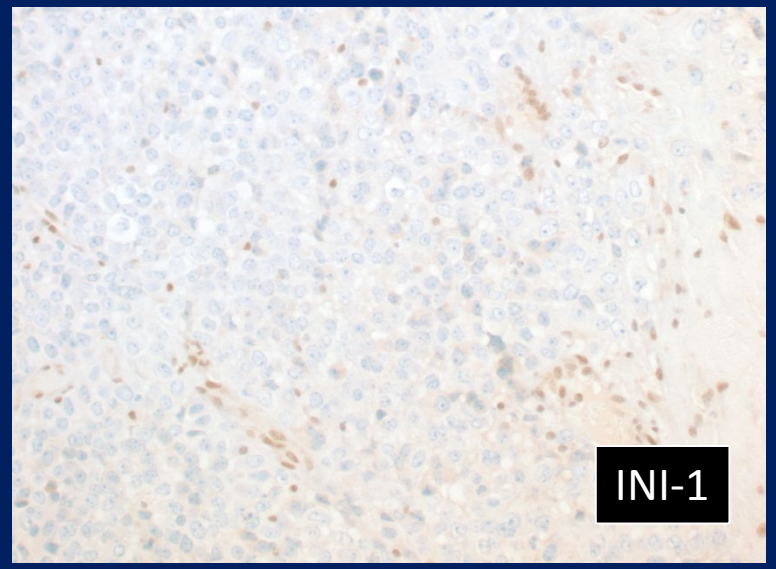


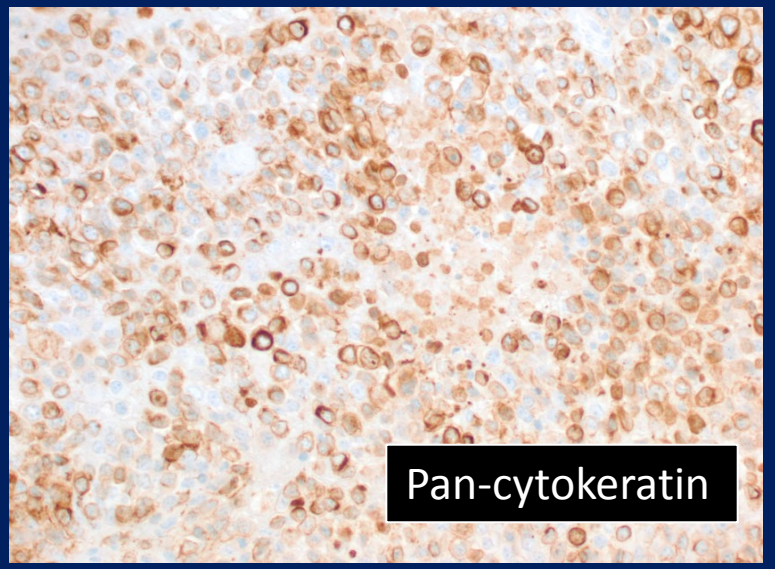
Differential?

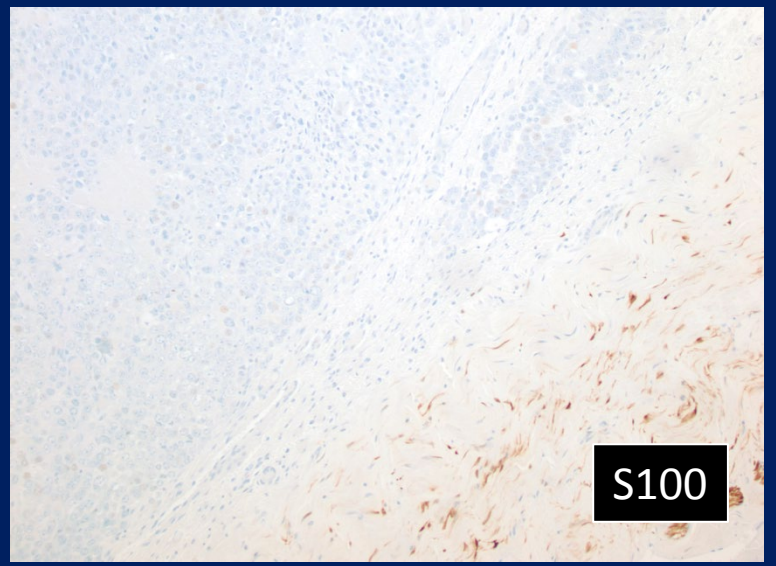
Differential Diagnoses

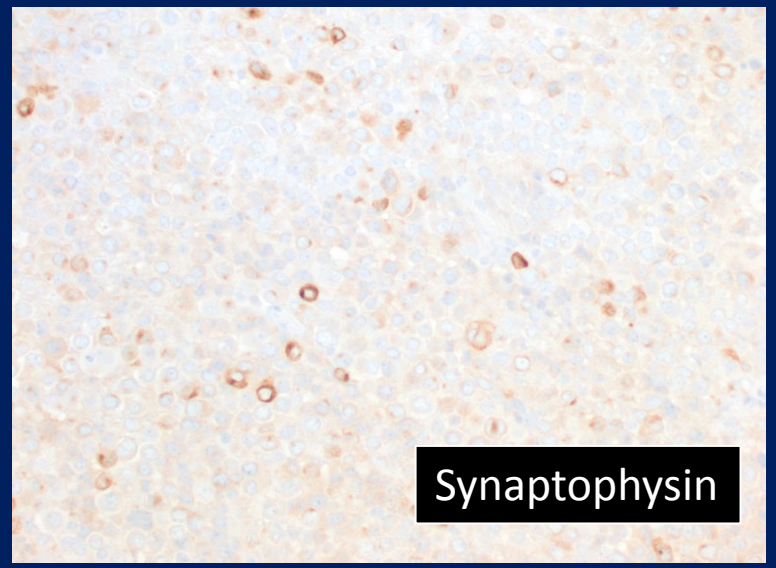
- Atypical Teratoid Rhabdoid Tumor (AT/RT)
- Epithelioid Malignant Peripheral Nerve Sheath Tumor
- Melanoma
- Rhabdomyosarcoma
- SMARCB1 (INI-1)-Deficient Sinonasal Carcinoma
- Olfactory Neuroblastoma
- Sinonasal Undifferentiated Carcinoma
- NUT Carcinoma
- Poorly differentiated chordoma
- High grade myoepithelial carcinoma
- Rhabdoid Meningioma

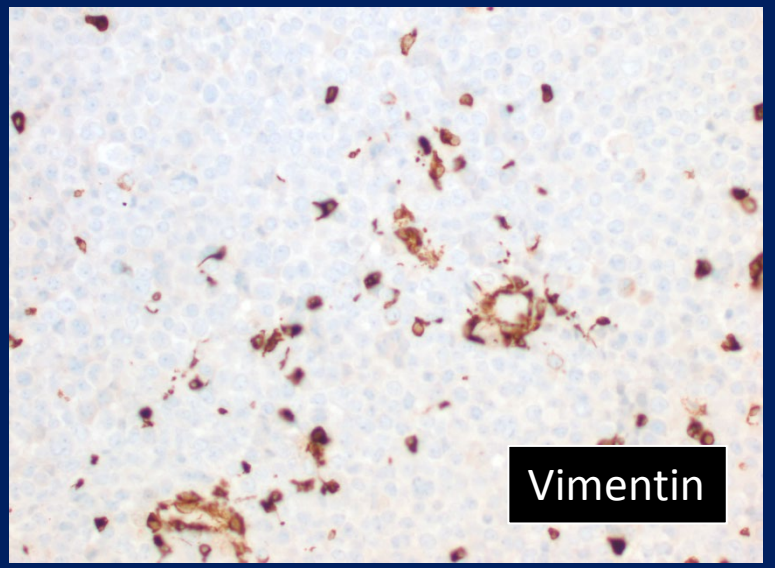








INI-1


Pan-cytokeratin


S100


Synaptophysin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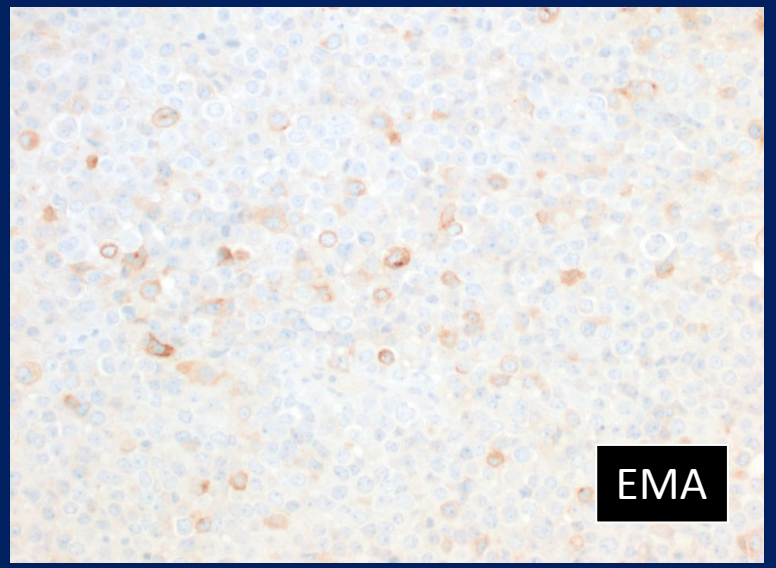
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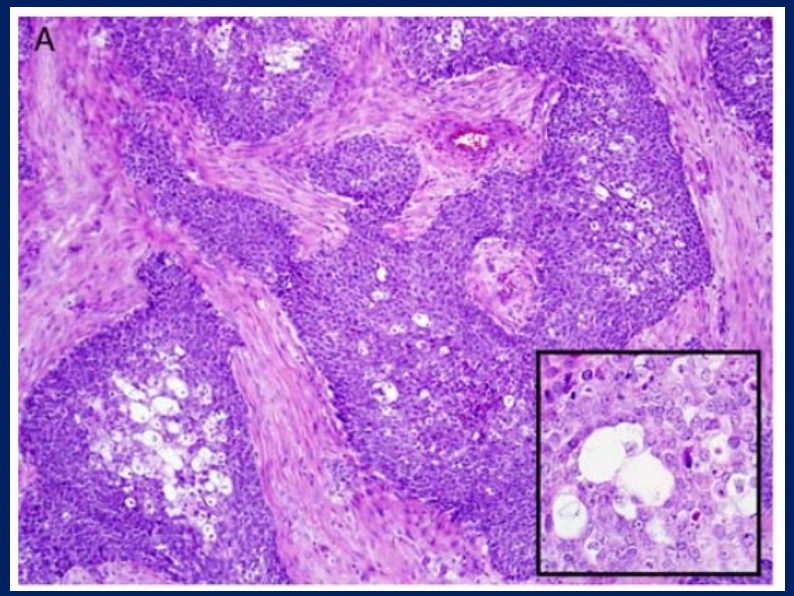
Diagnosis

- SMARCB1 (INI-1)-deficient sinonasal carcinoma

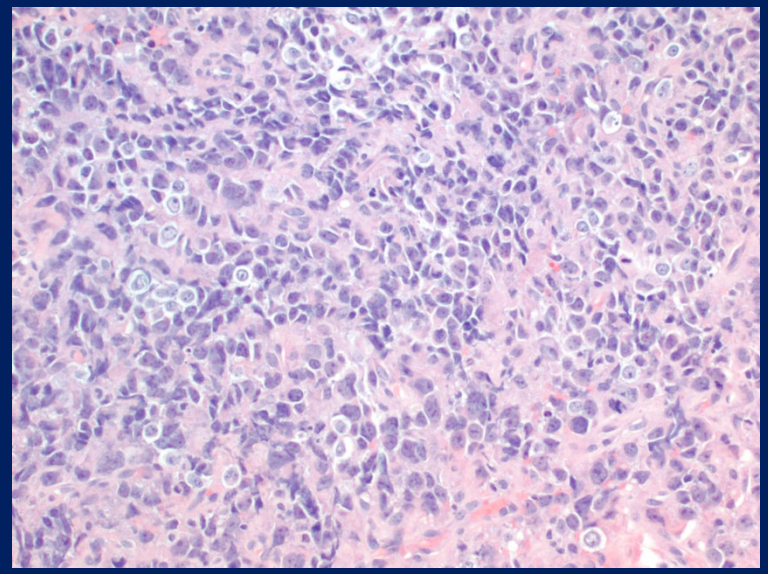
SMARCB1 (INI-1)-deficient Sinonasal Carcinoma

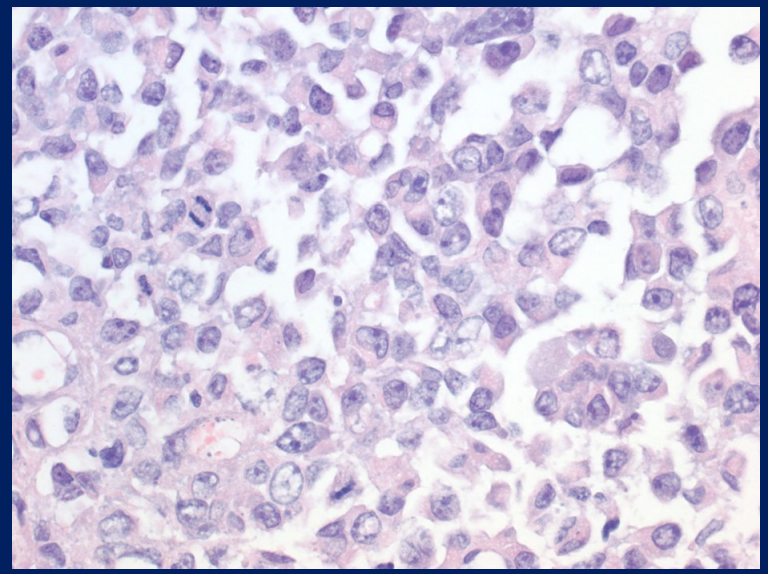
- Poorly differentiated sinonasal carcinoma
- Adults (19-87 years, Mean 52 years)
- Men and women are affected relatively equally
- Involves paranasal sinuses +/- nasal cavity
- Skull base involvement has been reported
- Poor prognosis, 54% of patients died between diagnosis and 102 months (median, 15 months)

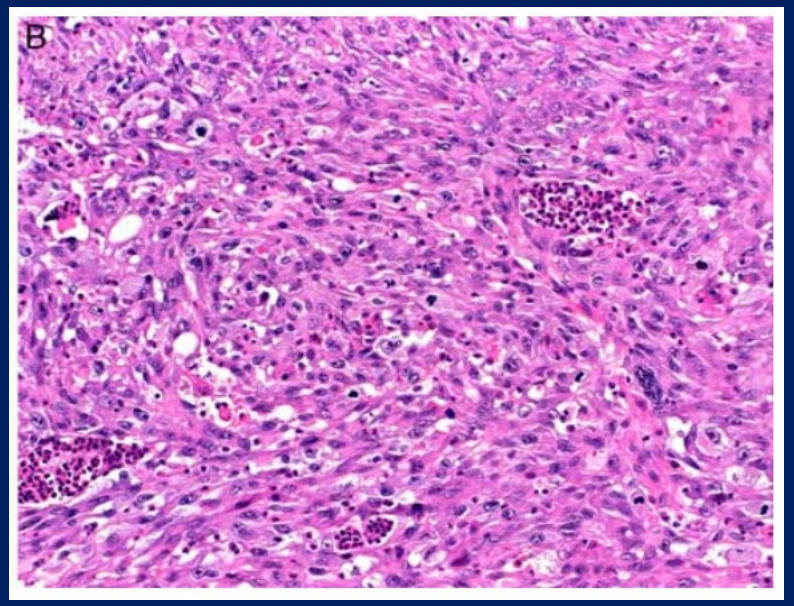
SMARCB1 (INI-1)-deficient Sinonasal Carcinoma



Agaimy et al. 2017



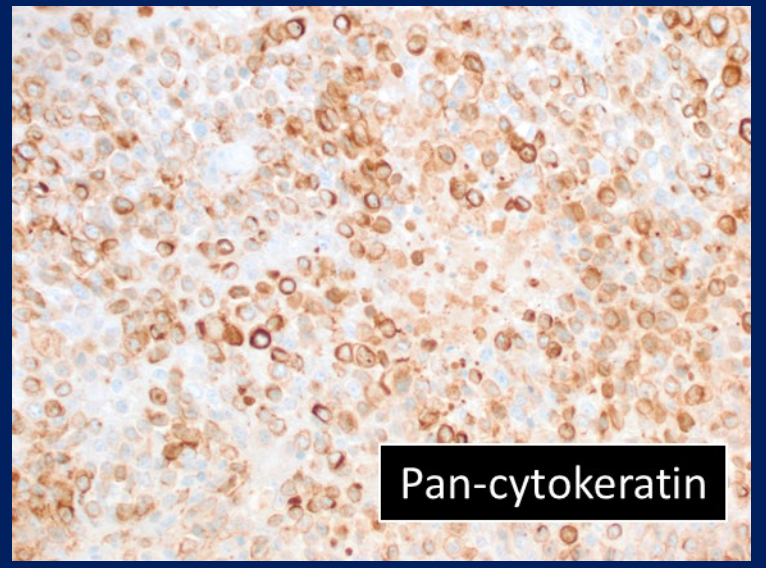


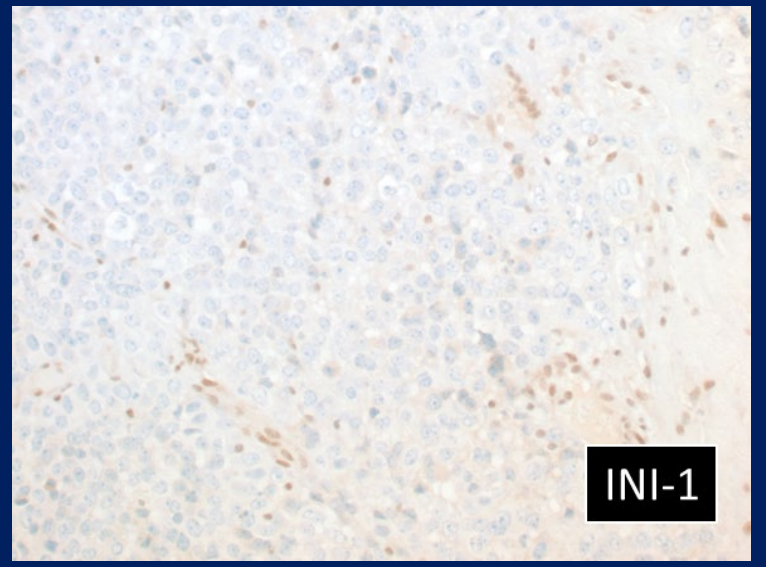


Agaimy et al. 2017

SMARCB1 (INI-1)-deficient Sinonasal Carcinoma

- Immunohistochemistry
  - SMARCB1 (INI-1) Loss
  - Positive
    - Pancytokeratin
    - CK5 (variable)
    - P63 (55%, diffuse positivity more common in basaloid histologic type)
    - P40 (44%)
    - Neuroendocrine markers (rarely)
    - May express P16 (not related to HPV)
  - Pertinent negatives
    - NUT-1
    - S-100
    - SOX10
    - Desmin
    - Myogenin
    - ERG
    - EBER
    - Hematopoeitic markers
    - CD99





SMARCB1 (INI-1)-deficient Sinonasal Carcinoma Differential

	SMARCB1 (INI-1)-deficient Sinonasal Carcinoma	AT/RT	Epithelioid MPNST	Rhabdomyosarcoma (Head and Neck)	Melanoma
Typical Demographics	Adult	Pediatric	Adult	Pediatric and Adult	Adult
Typical Location	Paranasal Sinuses	Intracranial	Extremities	Paranasal sinuses	Septum/Sinus
SMARCB1 (INI-1)	Loss	Loss	Loss	Retained	Retained
Pancytokeratin	Positive	Positive	Negative	Negative	Negative
P63	55% positive	Negative	Negative	Positive (cytoplasmic)	Negative
P40	44% positive	Negative	Negative	Negative	Negative
Neuroendocrine	Positive in subset	Positive	Negative	Negative	Negative
P16	Positive in subset	Negative	Positive in subset	Positive in subset	Positive
Desmin	Unknown	Negative	Negative	Positive	Negative
Myogenin	Unknown	Negative	Negative	Positive	Negative
S-100	Negative	Negative	Positive	Negative	Positive
SOX-10	Negative	Negative		Negative	Positive

Conclusion

- Important to consider SMARCB1 (INI-1)-deficient sinonasal carcinoma with lesional involvement of paranasal sinuses/skull base
- Diagnosis in the skull base depends on synthesis of histology, immunohistochemistry, imaging, and patient demographics
- Key histological clue is cellular monotony despite high grade appearance, necrosis, and mitoses

References

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Thank You