An unusual epithelioid neoplasm: Another mimic to add to the list!

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THE 47TH ANNUAL SCIENTIFIC MEETING

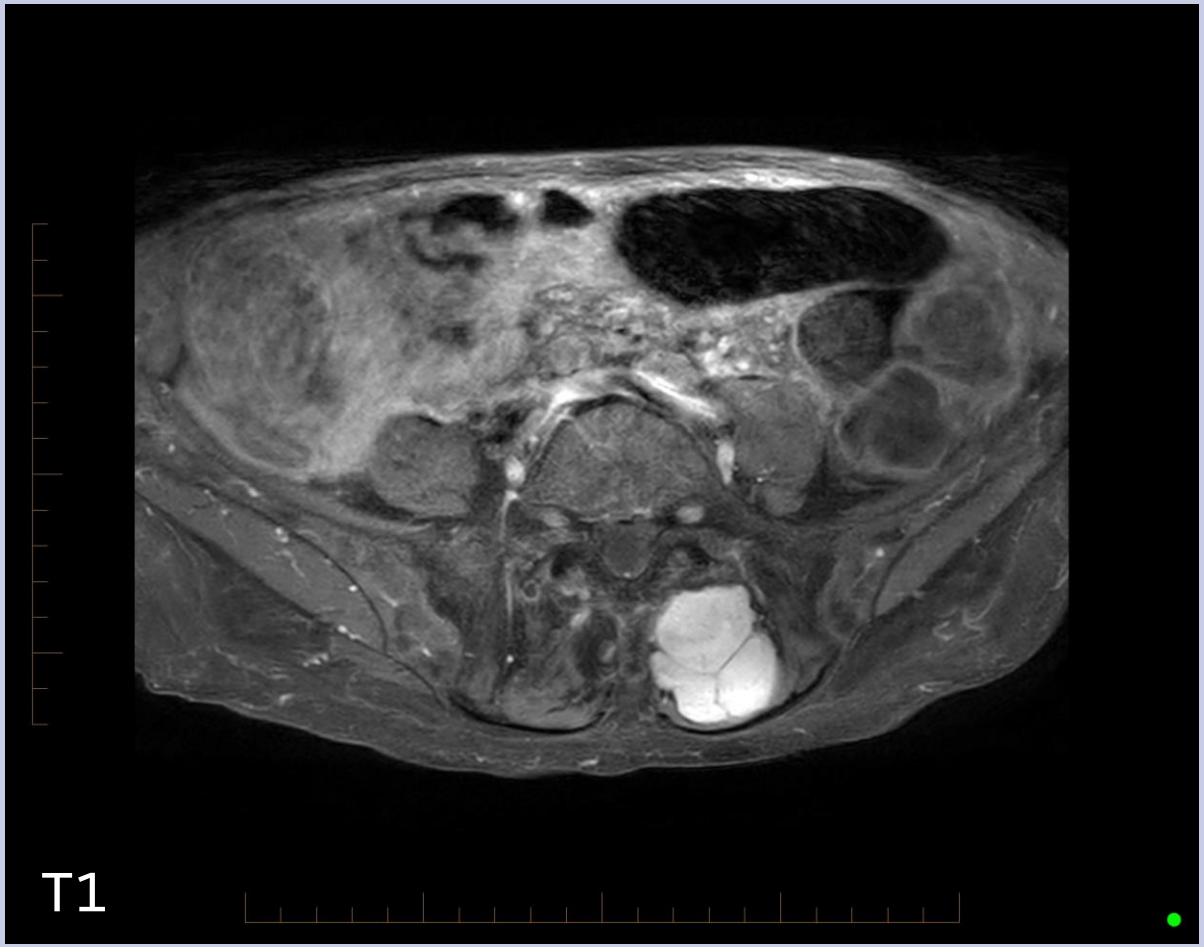
of the Australasian Division of the International Academy of Pathology

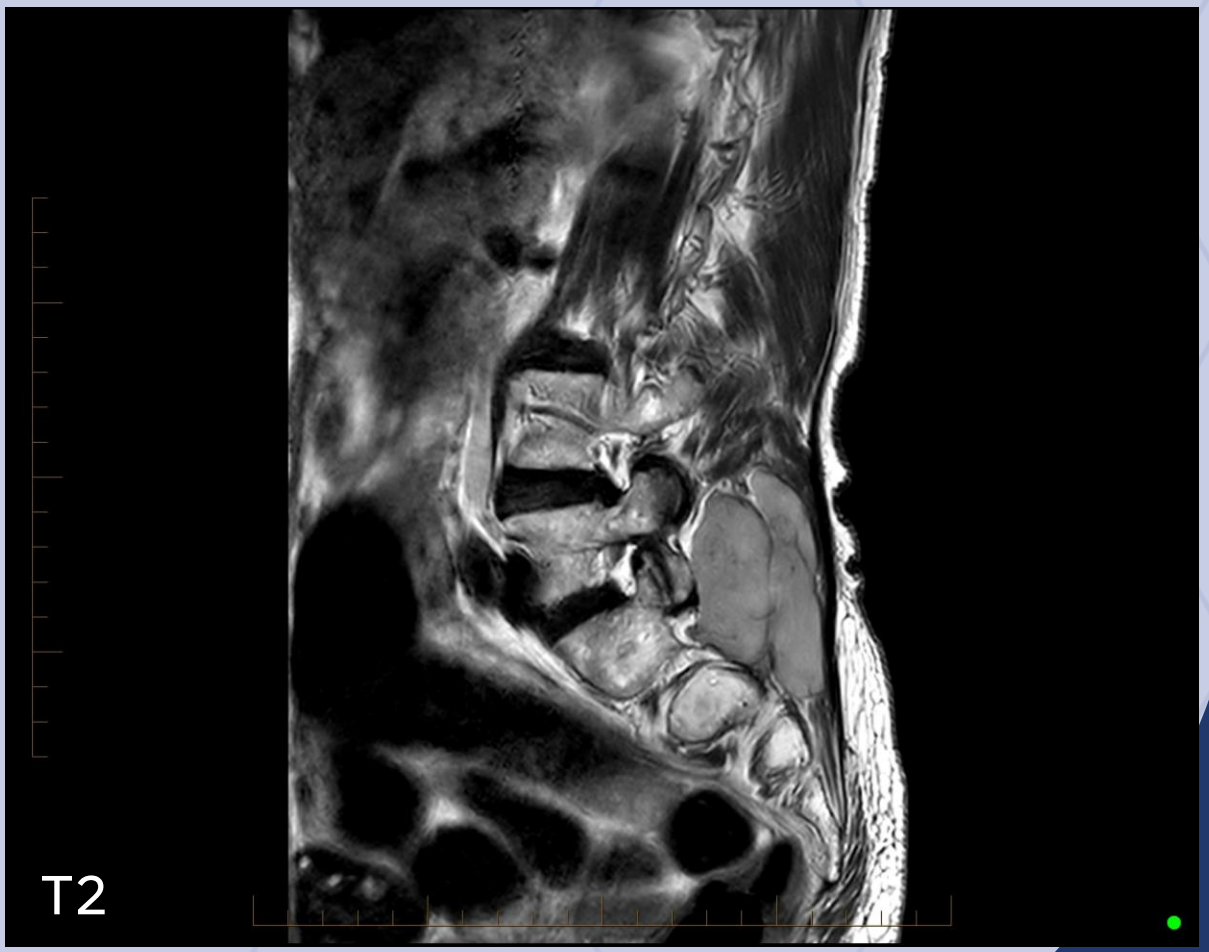
Disclosure of Relevant Financial Relationships

No relevant financial relationships.

Clinical presentation

- 90 year old woman
- Palpable painless mass of left lower back


T1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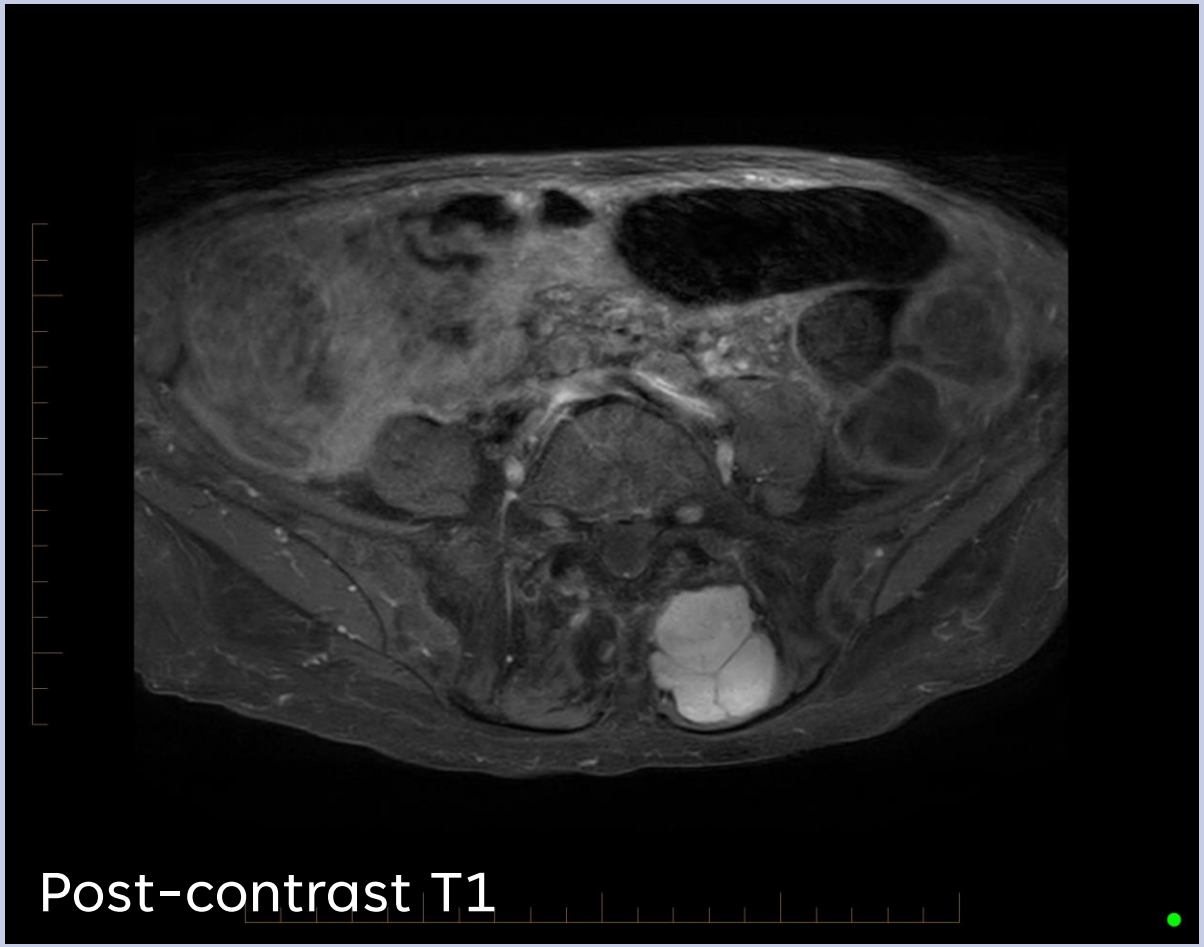
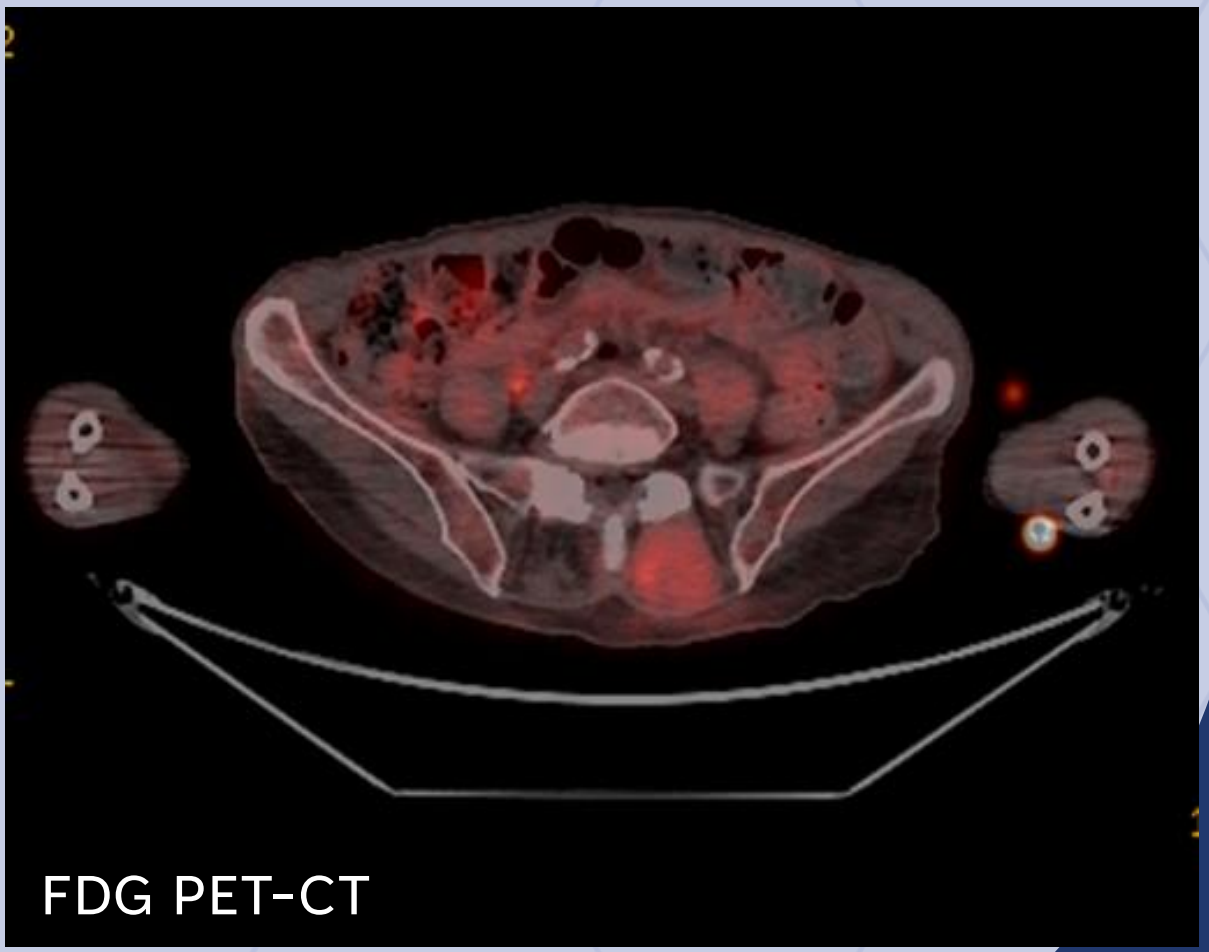
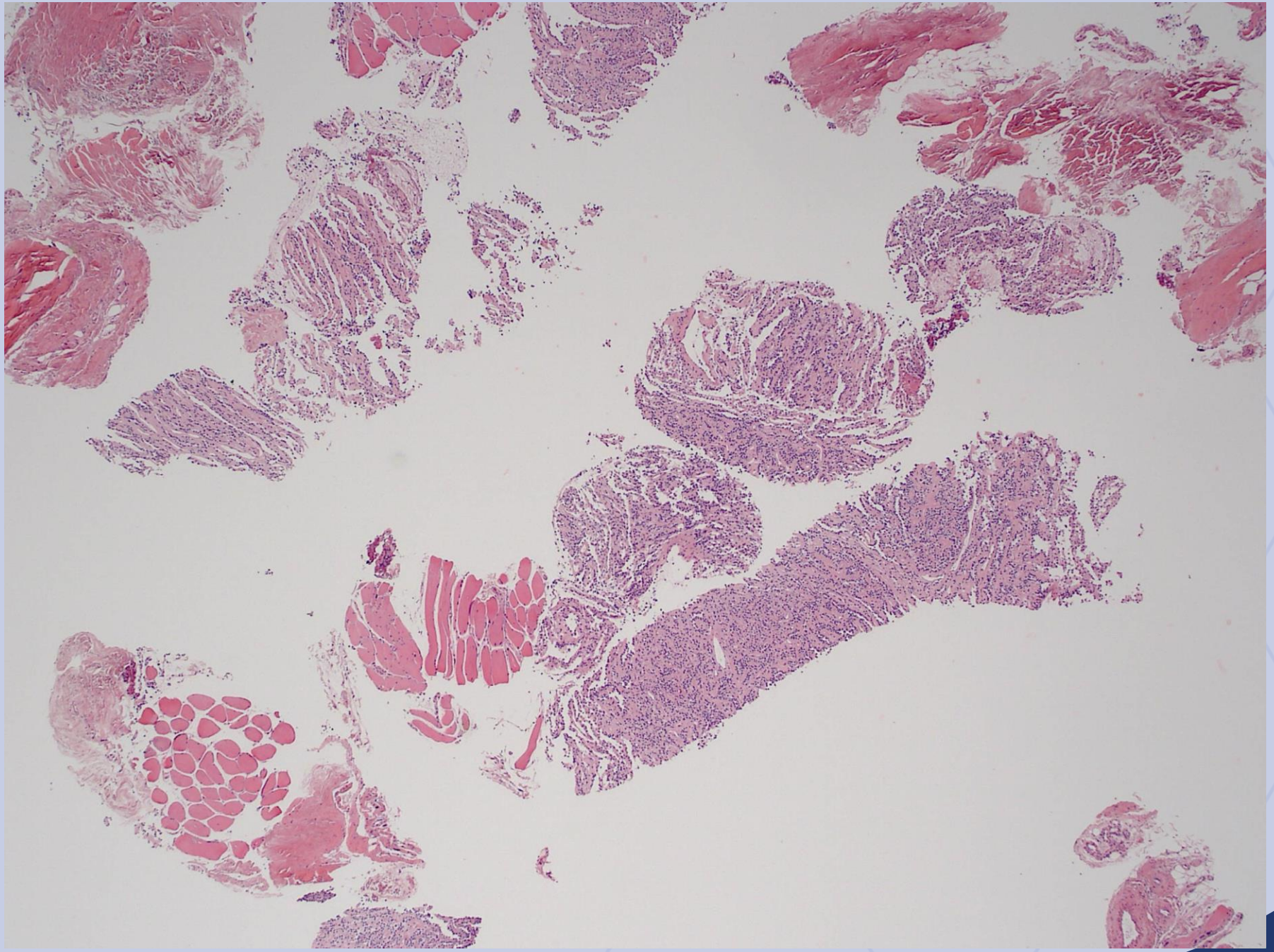
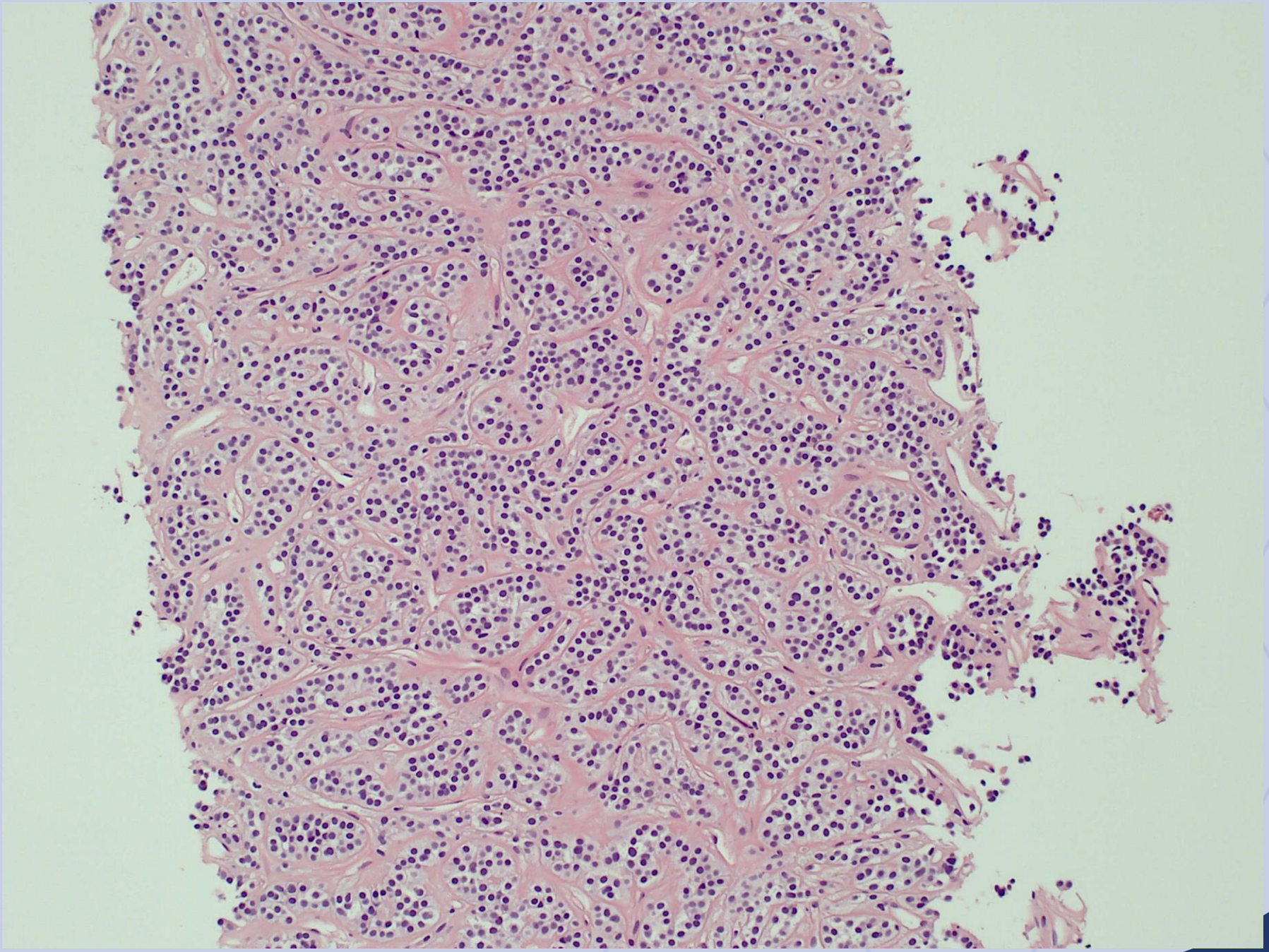


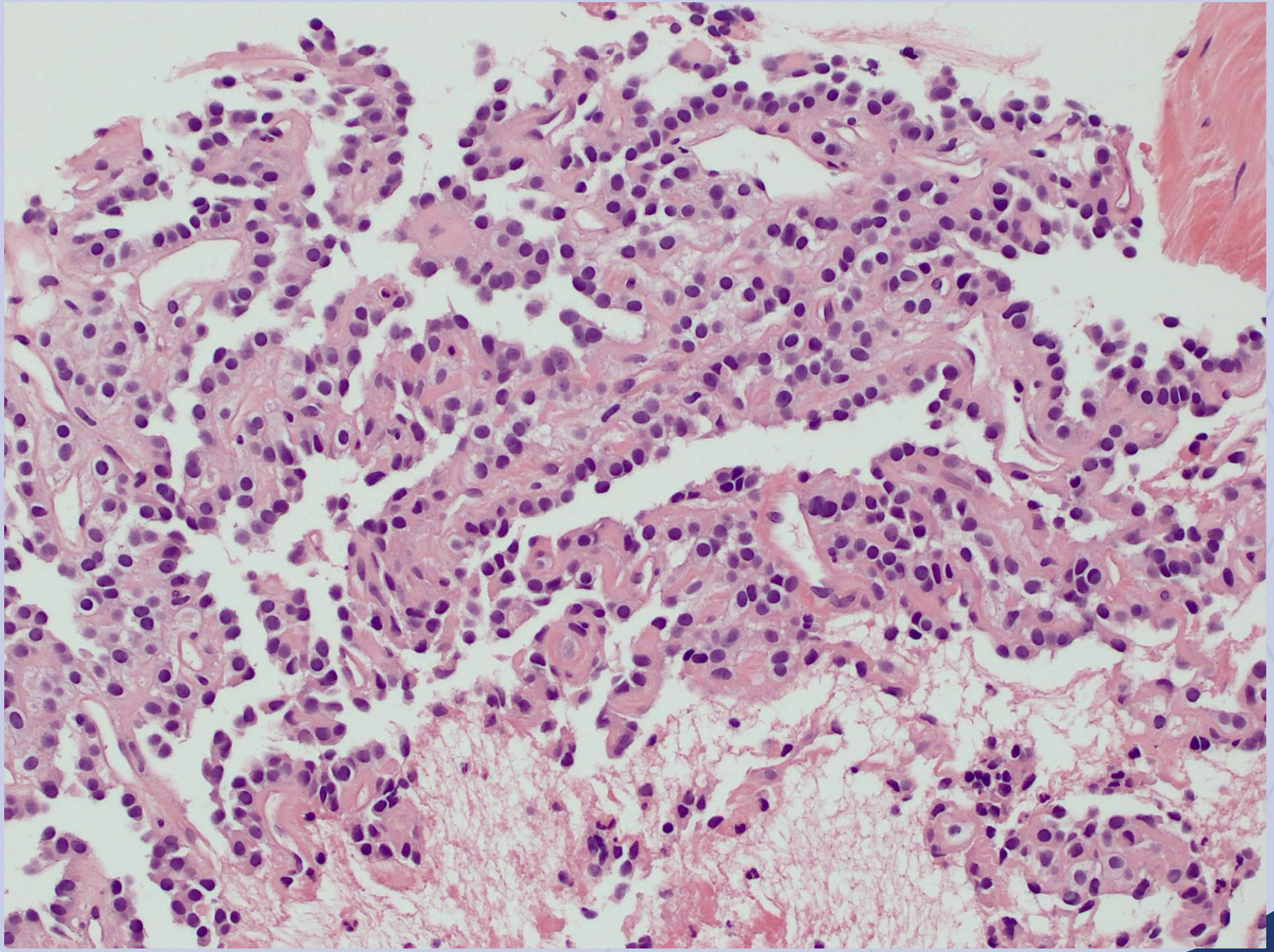
Post-contrast T1

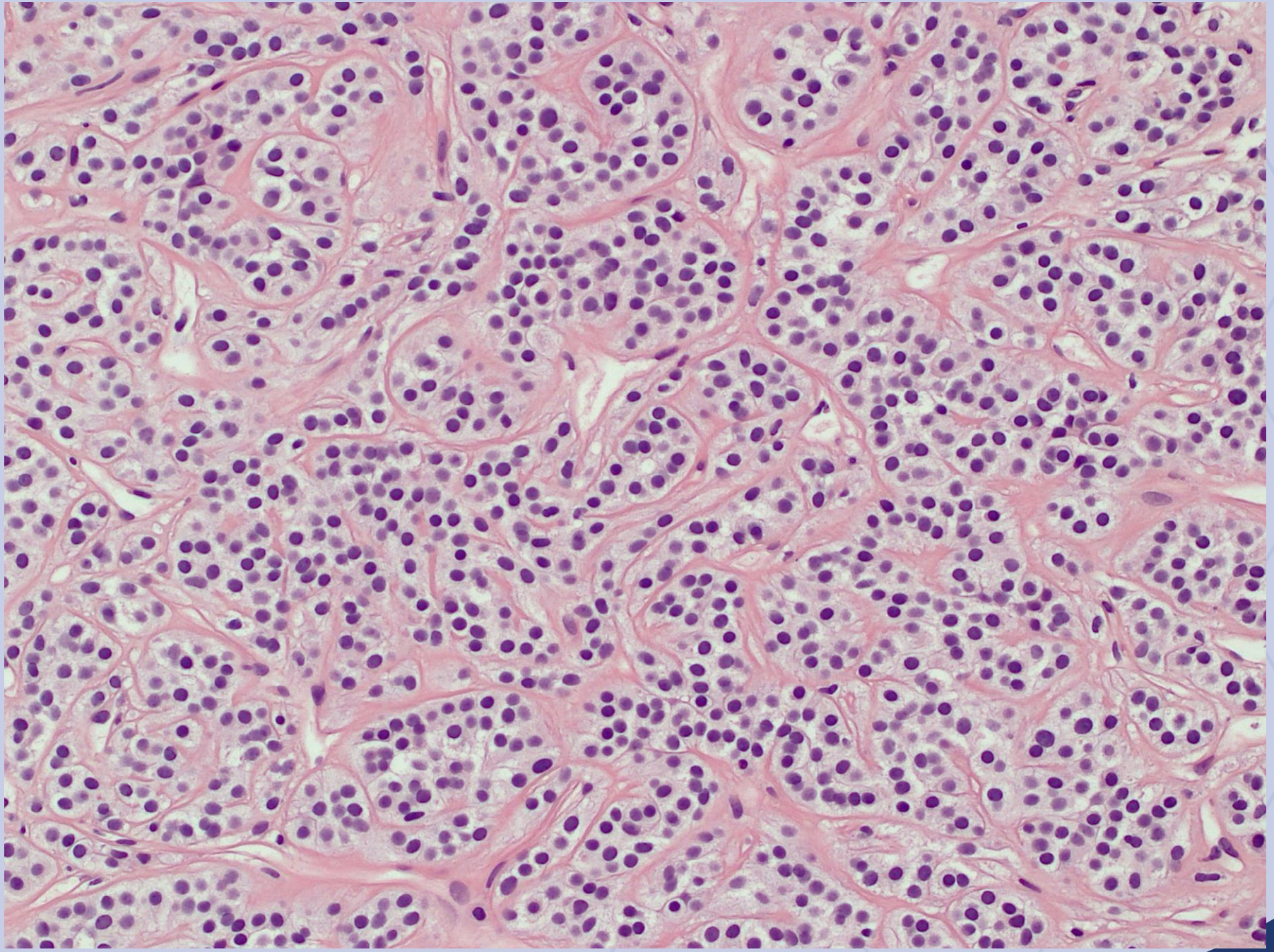


FDG PET-CT



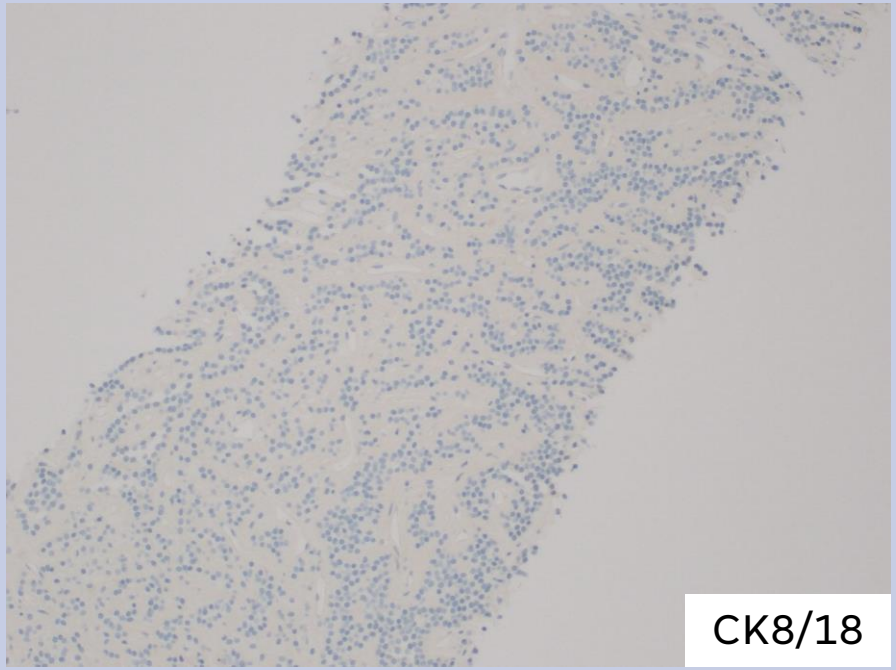




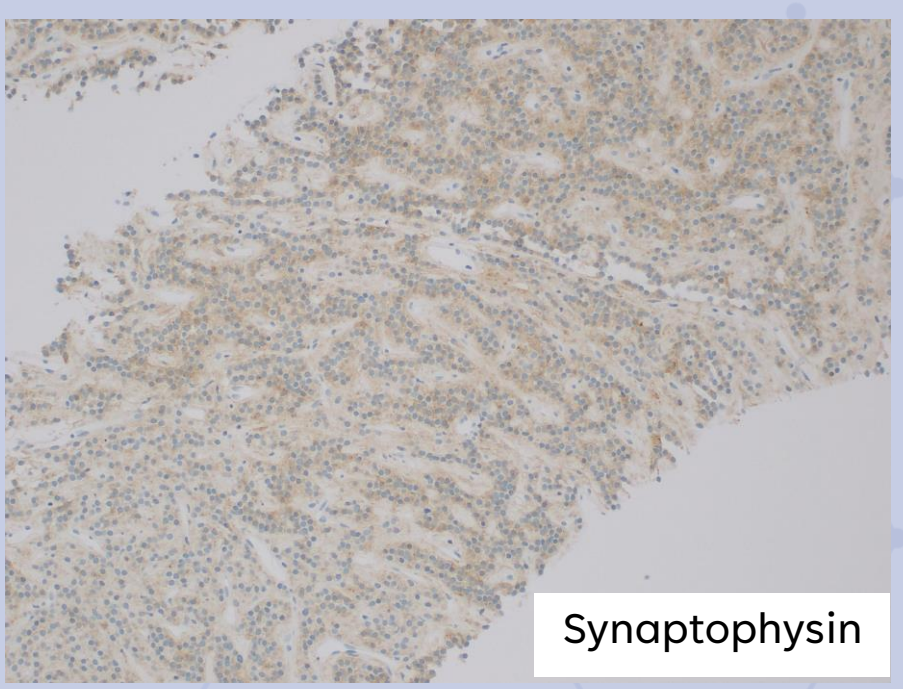


Negative markers

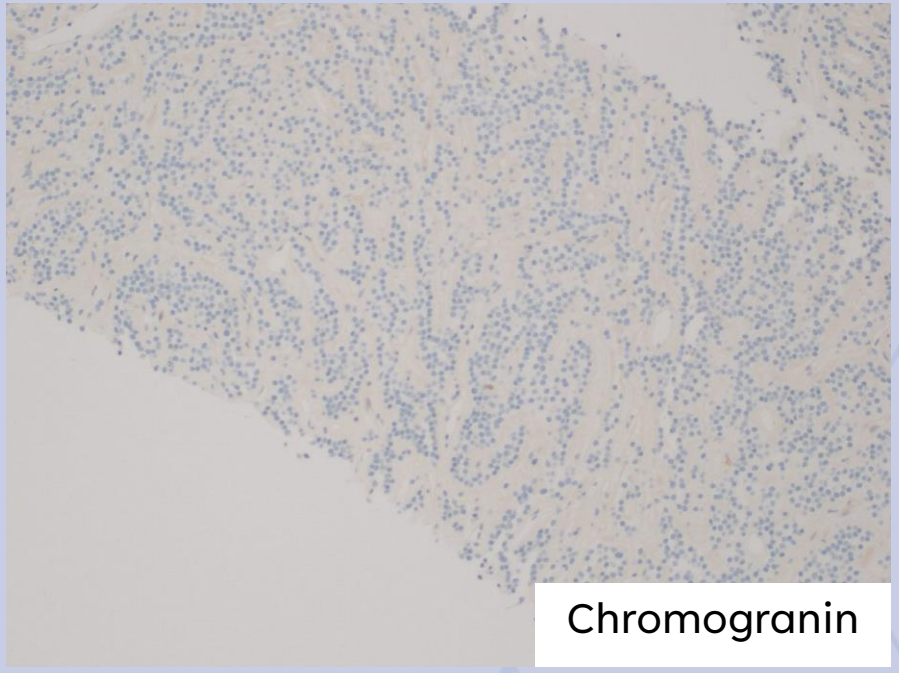
- EMA
- SMA
- h-Caldesmon
- Desmin
- S100 (F)
- Melan-A
- HMB45
- Inhibin
- CD34 (F)
- STAT6
- ERG
- CD117



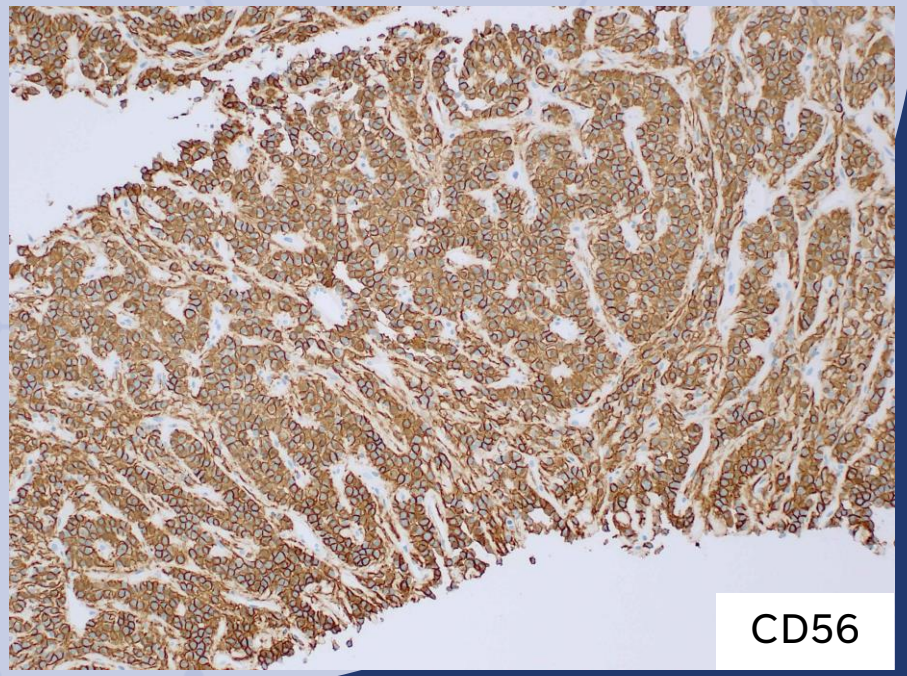
CK8/18



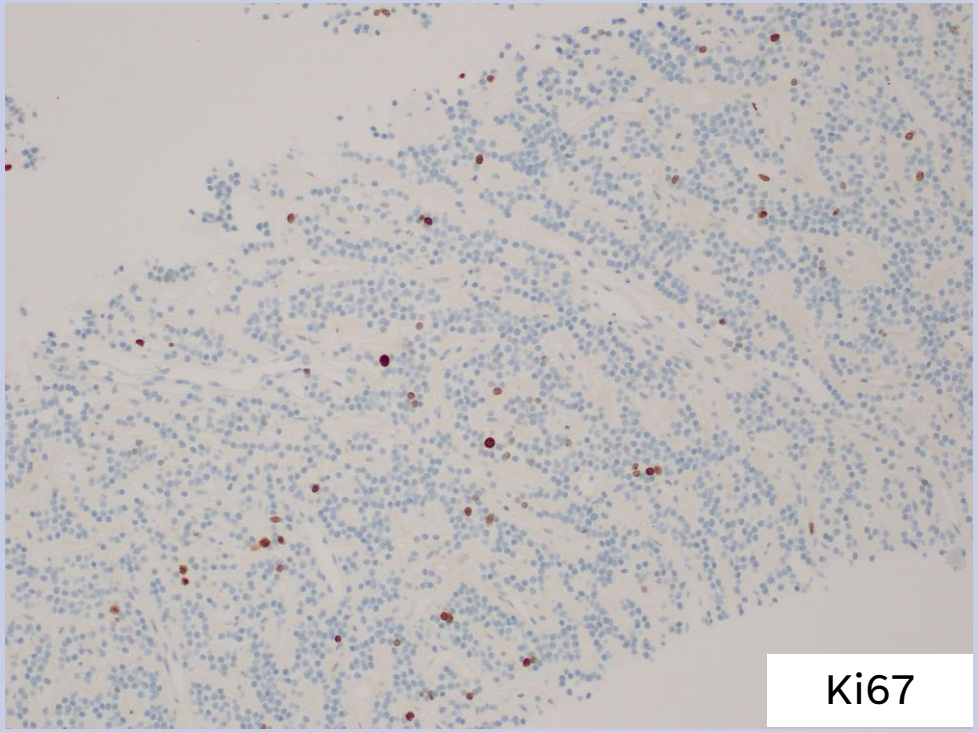
Synaptophysin



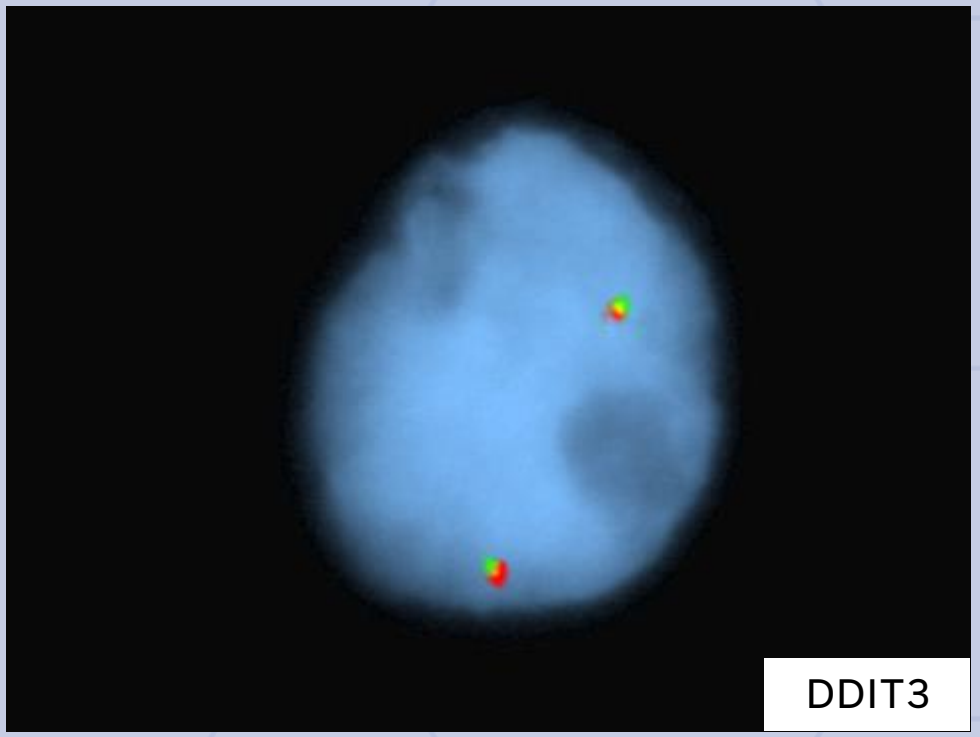
Chromogranin



CD56



Ki67



DDIT3

US-guided core biopsy, left lower lumbar paraspinal mass: **Low-grade epithelioid mesenchymal neoplasm, difficult to further subtype.**

Differential diagnoses - carcinoid tumour, paraganglioma, glomus tumour, PEComa, ependymoma and GLI1-rearranged malignant epithelioid neoplasm.

ORIGINAL ARTICLE

Pseudoendocrine Sarcoma

Clinicopathologic Analysis of 23 Cases of a Distinctive Soft Tissue Neoplasm With Metastatic Potential, Recurrent CTNNB1 Mutations, and a Predilection for Truncal Locations

David J. Papke Jr, MD, PhD, Brendan C. Dickson, MD, MSc,†‡ Lynette Sholl, MD,* and Christopher D.M. Fletcher, MD, FRCPath**

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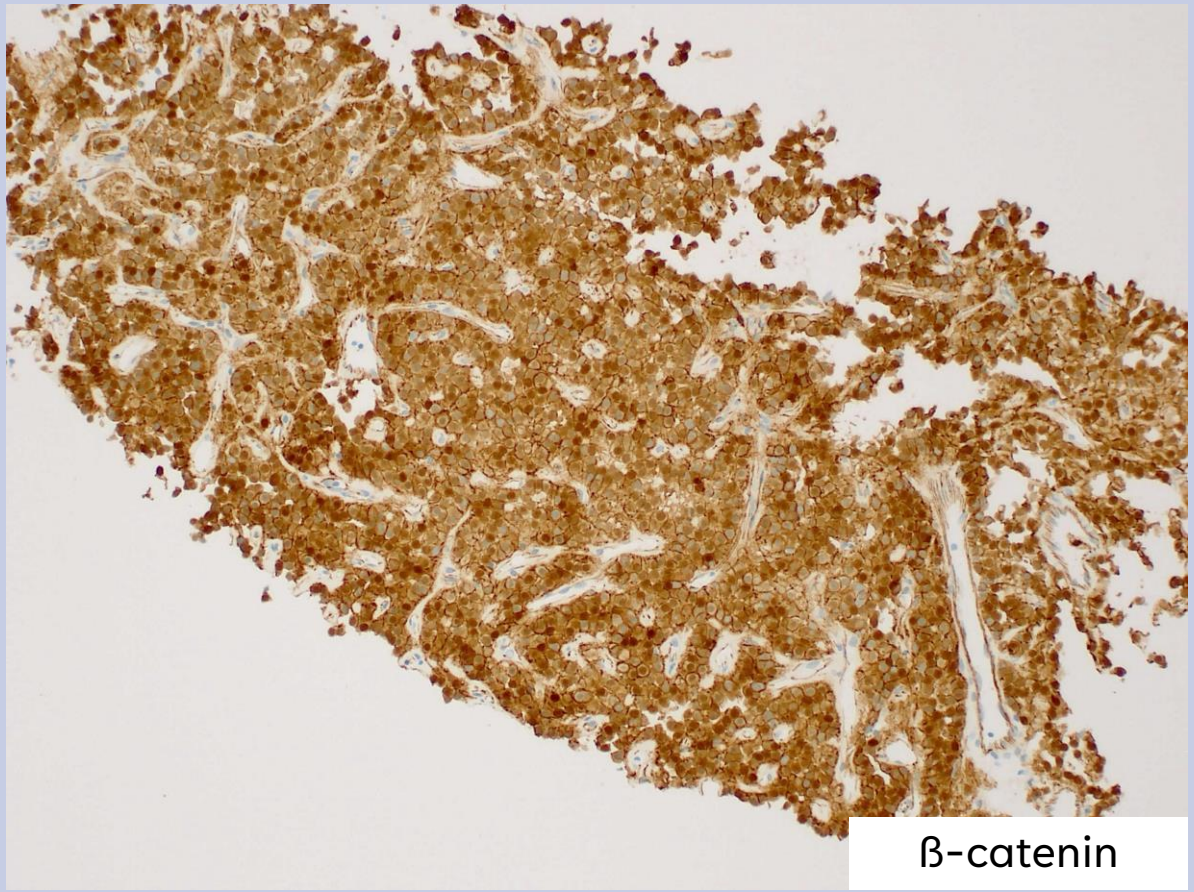


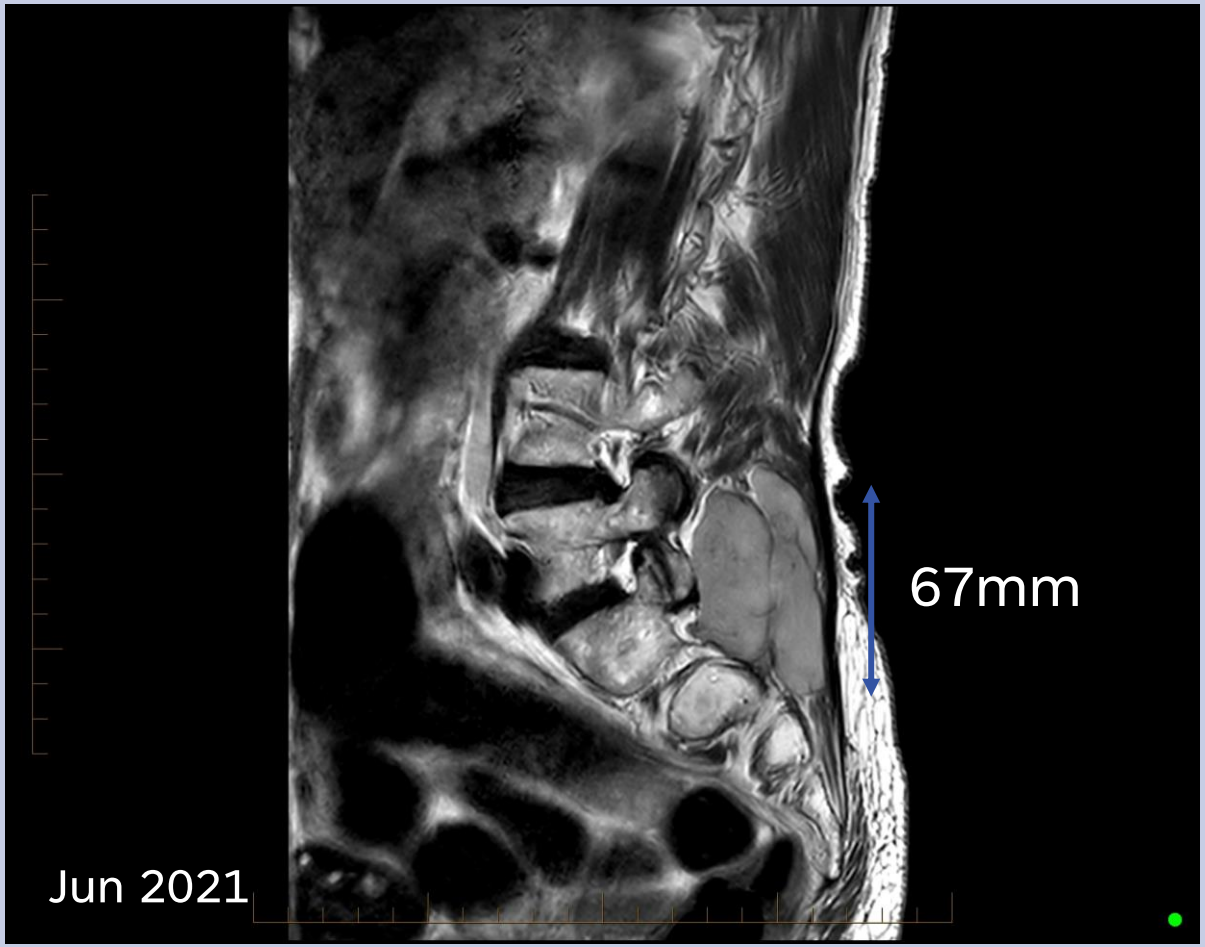
ß-catenin

Molecular testing

- Illumina Ampliseq 33 gene NGS assay
- ***CTNNB1*** **p.(ser33Phe) 43% VAF**

US-guided core biopsy, left lower lumbar paraspinal mass: Epithelioid mesenchymal neoplasm with *CTNNB1* mutation, consistent with so-called

Pseudoendocrine sarcoma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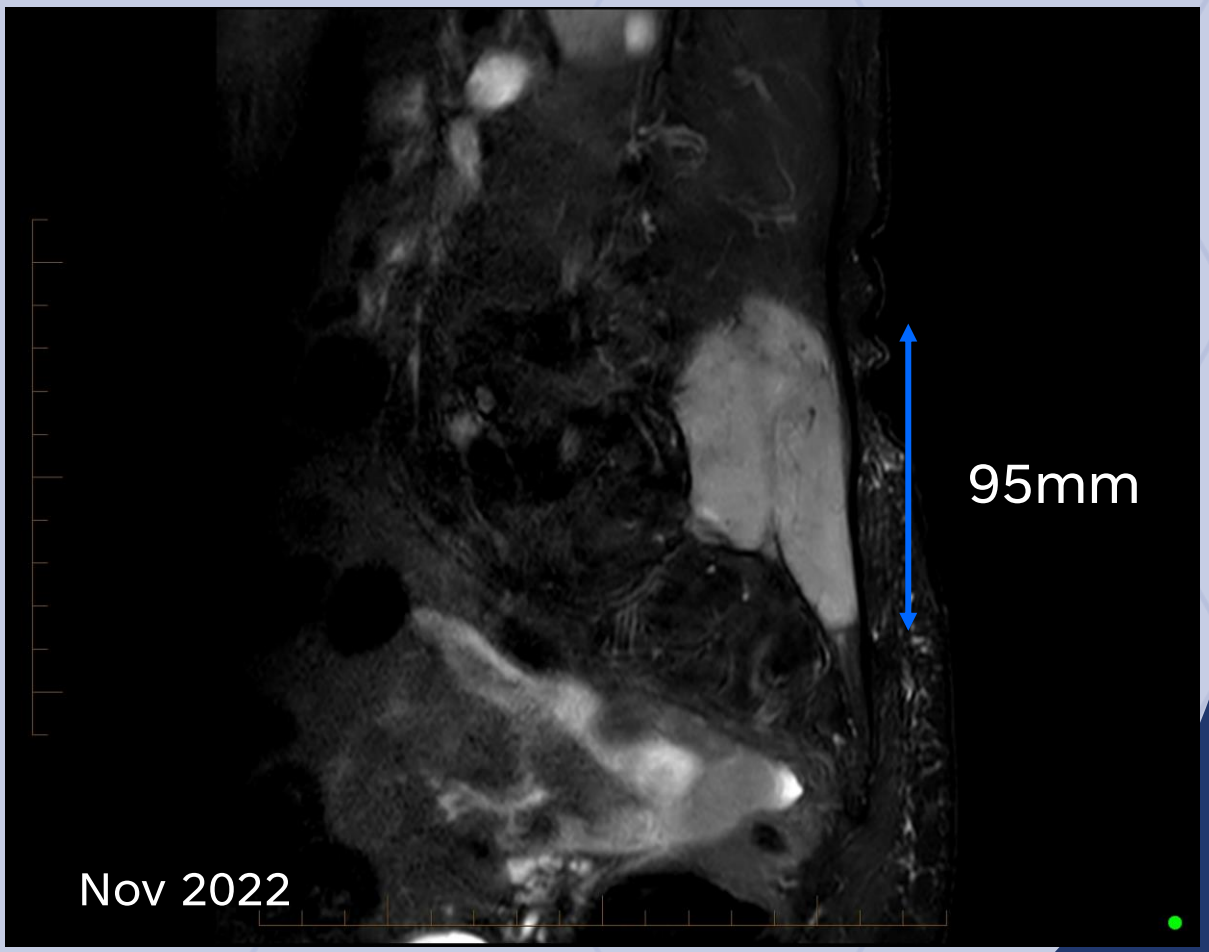
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Virchows Archiv
https://doi.org/10.1007/s00428-022-03476-4

ORIGINAL ARTICLE

Check for updates

Case report: pseudoendocrine sarcoma, a clinicopathologic report of a newly described soft tissue neoplasm

Elena Bellan[1,2] · Francesca Zanco[3] · Francesca Baciorri[3] · Luisa Toffolatti[3] · Angelo P. Dei Tos[1,2] · Marta Sbaraglia[1,2]

LETTER TO THE EDITOR

Meningioma-like Ultrastructural Features of Pseudoendocrine Sarcoma

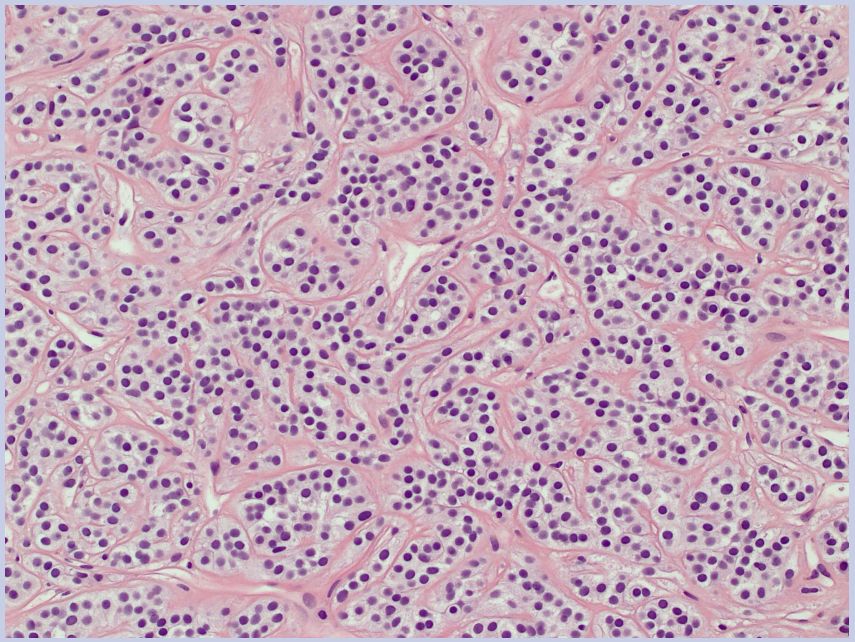
To the Editor:

Pseudoendocrine sarcoma, a provisional term for a rare distinctive and recently described tumor, most commonly involves the paravertebral soft tissue of older individuals.[1] Microscopically, pseudoendocrine sarcoma exhibits a nested and trabecular architecture and is composed of uniform epithelioid cells, with round nuclei and speckled chromatin reminiscent of well-differentiated neuroendocrine tumors.[1] However, pseudoendocrine sarcoma lacks the expression of neuroendocrine immunohistochemical markers, including synaptophysin, chromogranin, and INSM1. In addition, most tumors are negative for epithelial markers, including

Pseudoendocrine sarcoma (n=26)

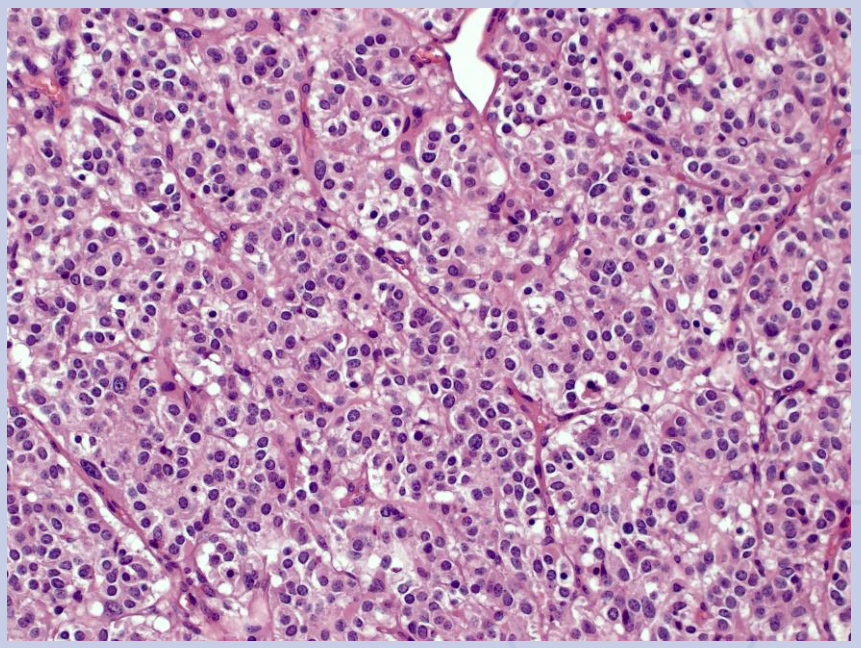
- 15 (58%) males : 11 females
- Median age 62 years (29 – 90)
- 85% truncal locations (esp. paravertebral soft tissues)
  - Posterior head, thigh, orbit
  - All deep soft tissue (intramuscular), some involving bone (vertebrae)
- Palpable (painful) mass, others incidental

Differential diagnosis



PSEUDOENDOCRINE SARCOMA

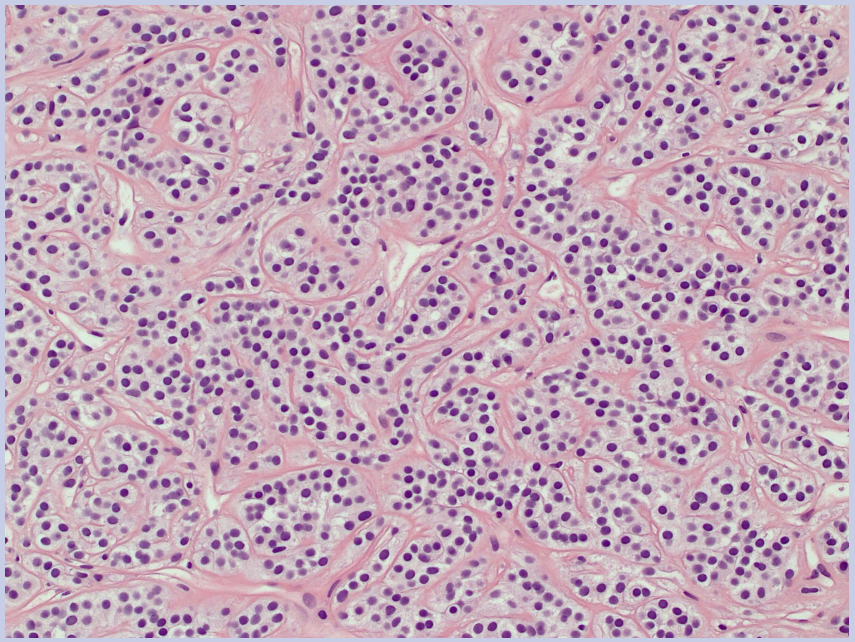
- Negative for keratins and neuroendocrine markers
- Nuclear B-catenin and *CTNNB1* mutation



WD NEUROENDOCRINE TUMOUR

- Stains for keratins and neuroendocrine markers
- No nuclear B-catenin or *CTNNB1* mutation

Differential diagnosis



PSEUDOENDOCRINE SARCOMA

- Negative for neuroendocrine markers
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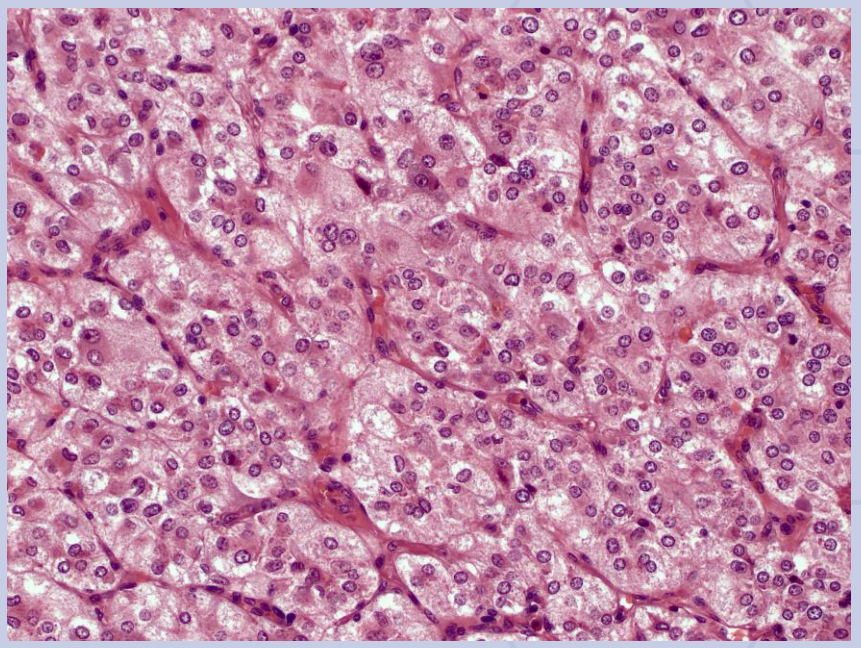
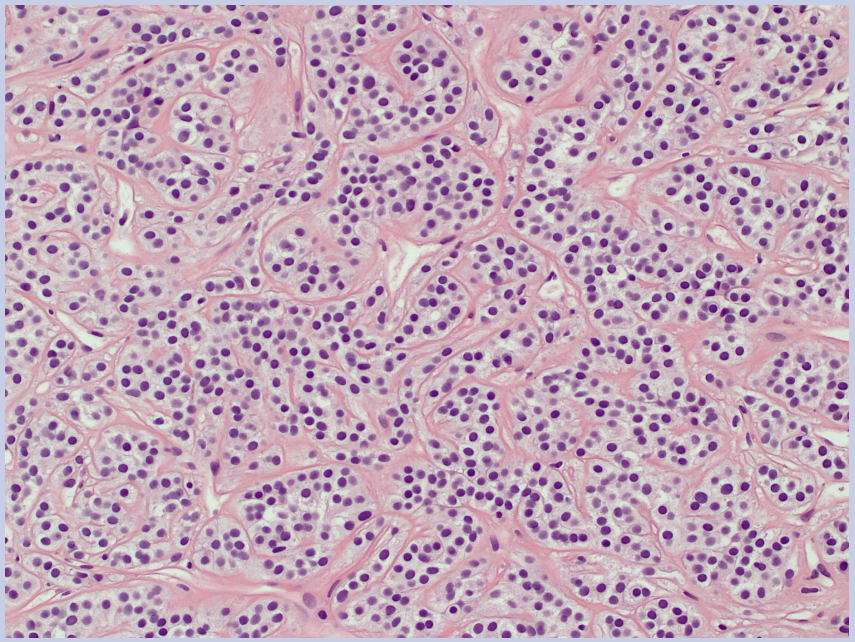


PARAGANGLIOMA

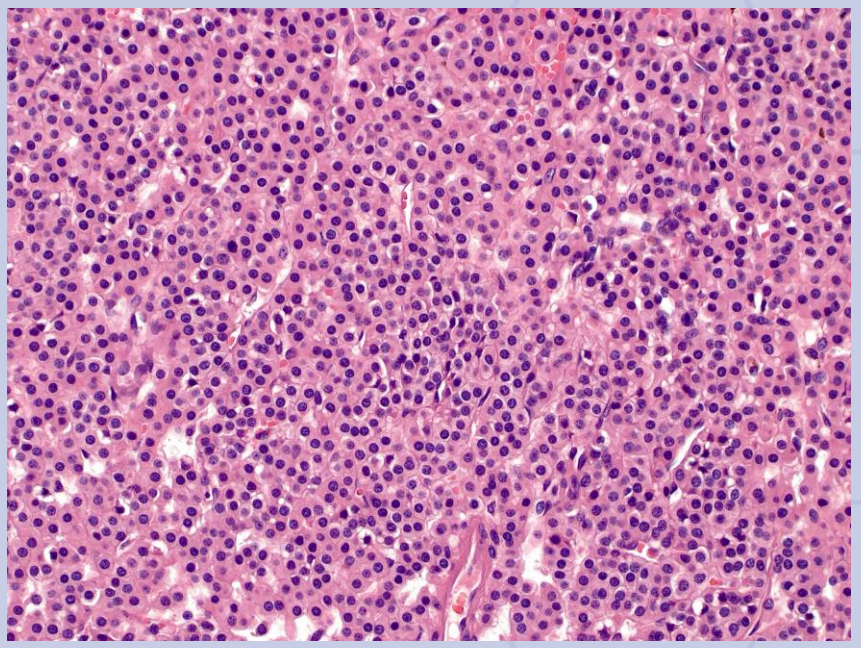
- Stains for neuroendocrine markers and SOX10 (SC)
- *SDH, NF1, RET, VHL* alterations

Differential diagnosis



PSEUDOENDOCRINE SARCOMA

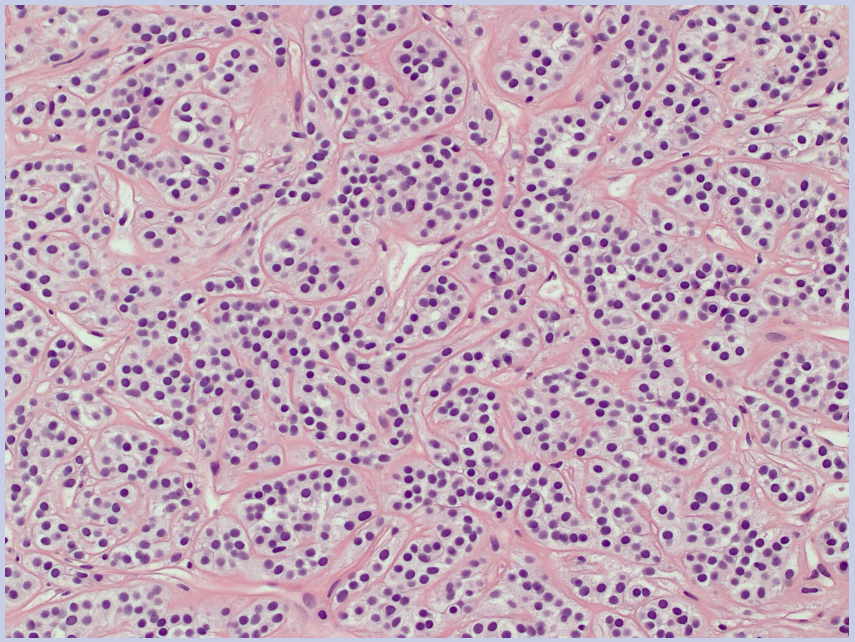
- Nuclear B-catenin and *CTNNB1* mutation



GLOMUS TUMOUR

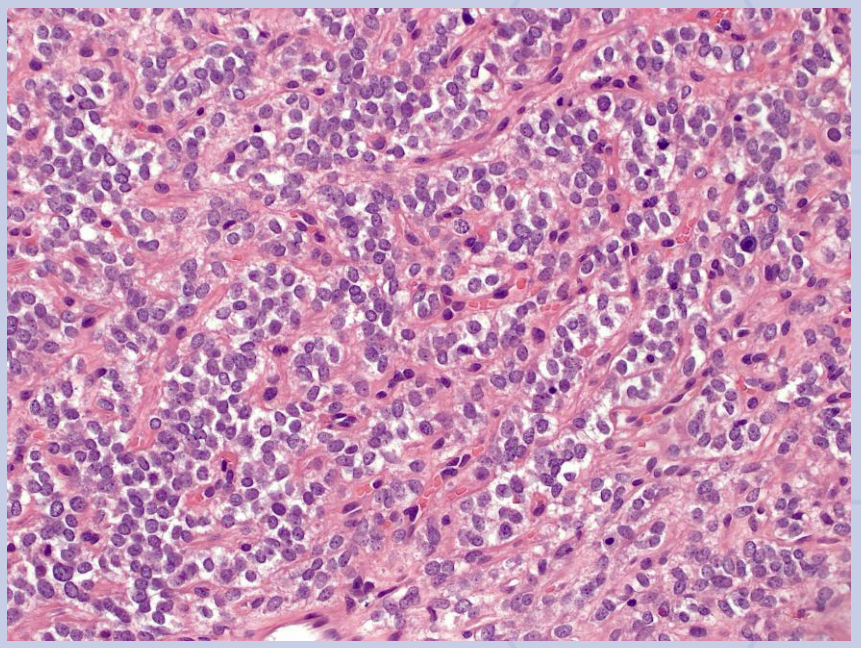
- Prominent cell borders
- Stains for SMA and h-caldesmon

Differential diagnosis



PSEUDOENDOCRINE SARCOMA

- Monotonous and purely epithelioid
- Nuclear B-catenin and *CTNNB1* mutation



GLI1-m. EPITHELIOID TUMOUR

- Cytological variability, with spindle cells
- GLI1 (IHC and/or molecular)

Pseudoendocrine sarcoma (n=17)

- Median follow-up 3.5 years (2mth – 20 yrs)
- 45% local recurrence at 3 – 6 years
  - Most with positive margins
- 20% distant metastases at 1 – 20 years
  - Lymph node and lungs
  - One patient AWED at 9 years
- No recorded death from disease
- Recommended treatment – wide excision

Pseudoendocrine sarcoma

- Distinct mesenchymal neoplasm of uncertain differentiation
- Older adults at truncal locations
- Nested epithelioid morphology resembling neuroendocrine tumours
- Negative for keratins and neuroendocrine markers; strong and diffuse nuclear B-catenin
- *CTNNB1* mutation
- Significant risk of recurrence and metastases

Acknowledgements

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