SELNET Virtual MDT

27th of February 2025

Host institution: Hospital Clinico san Carlos, Madrid, Spain

SaludMadrid Hospital Clínico San Carlos

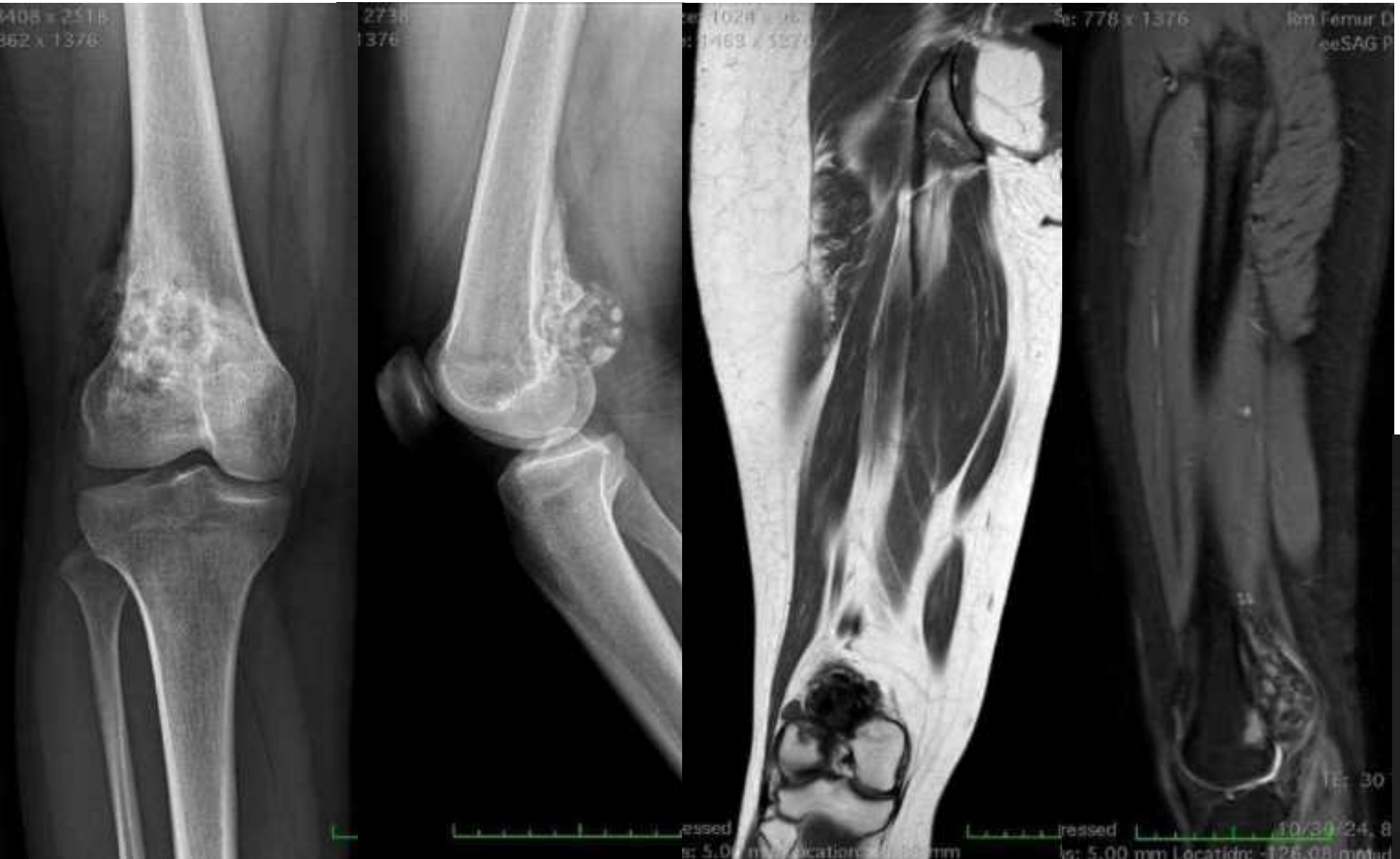
European Reference Network for rare or low prevalence complex diseases

Network Adult Cancers

Patient #1 Panamá

- 25-year-old female
- No previous history of disease
- One-year history of pain and discomfort in the posterior knee. Worsens with flexion over 90°
- On physical exam
  - Palpable mass on popliteal region of the right knee, normal rango of motion.
  - Pain with knee hyperflexion

Imaging studies



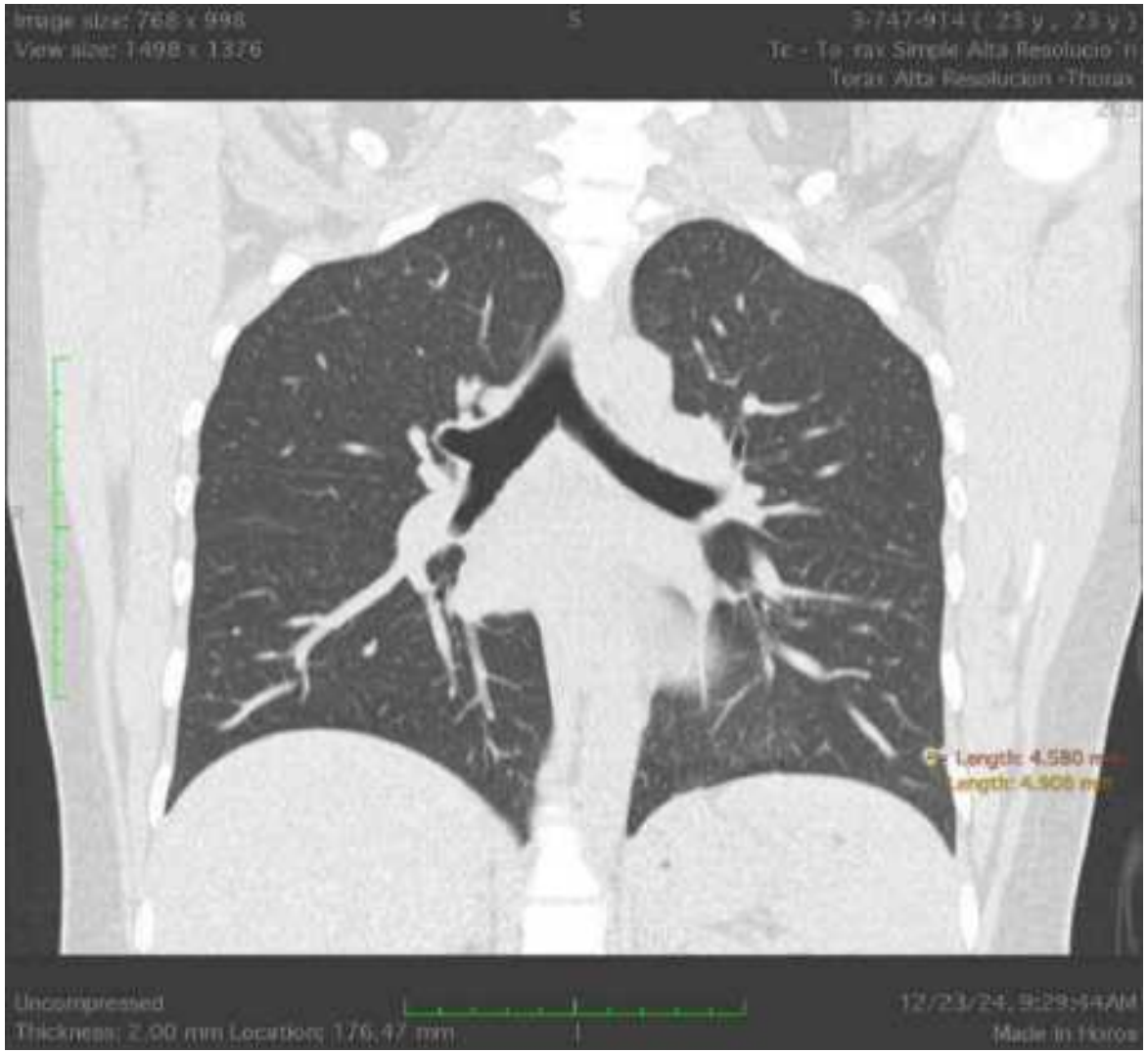


Image size: 768 x 998
View size: 1498 x 1376
S
3-747-914 (23 y , 23 y)
Tc - To rax Simple Alta Resolucio n
Toras Alta Resolucion -Thorax
Length: 4.580 mm
Length: 4.508 mm
Uncompressed
Thickness: 2.00 mm Location: 176.47 mm
12/23/24, 9:29:44AM
Made in Horos

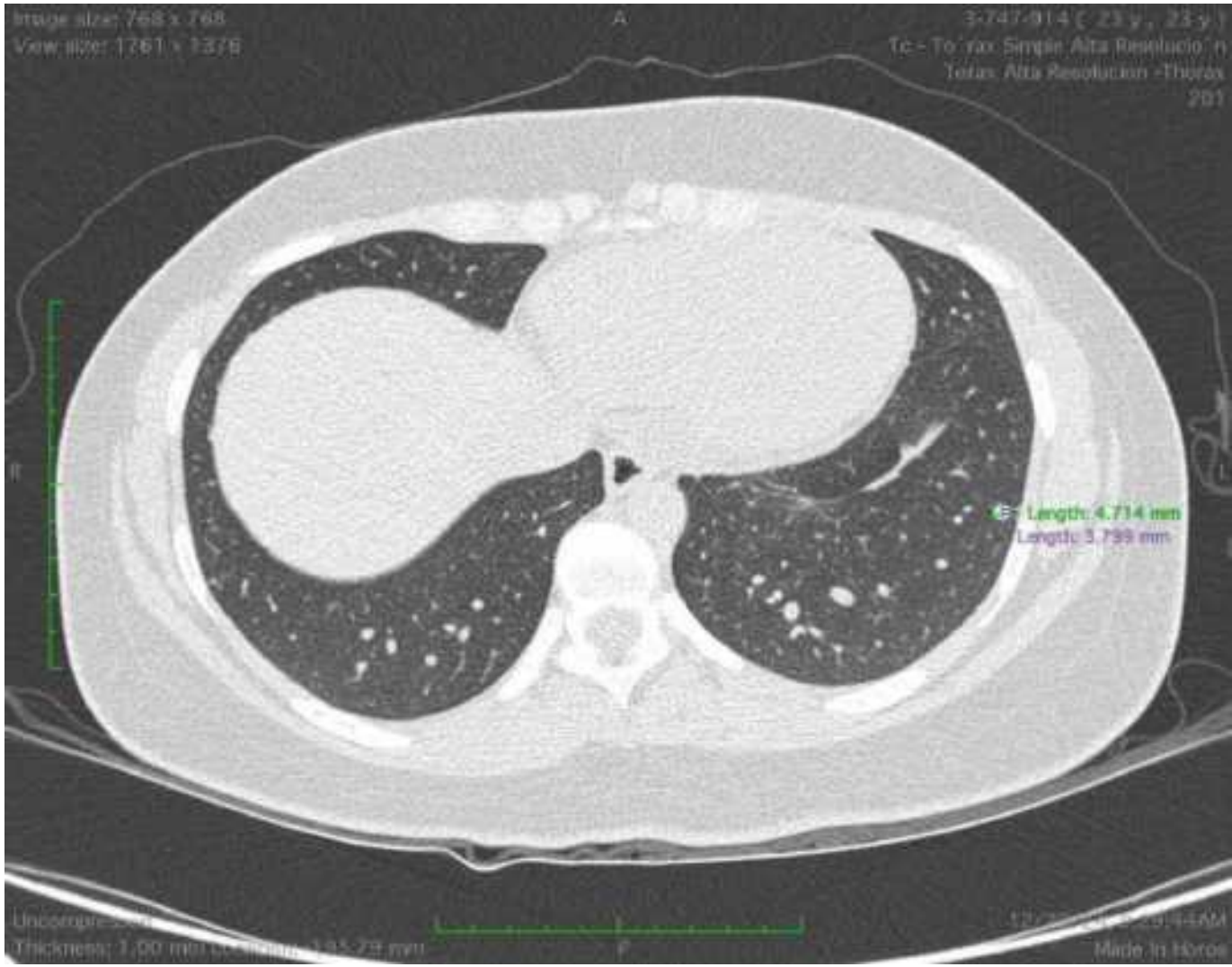
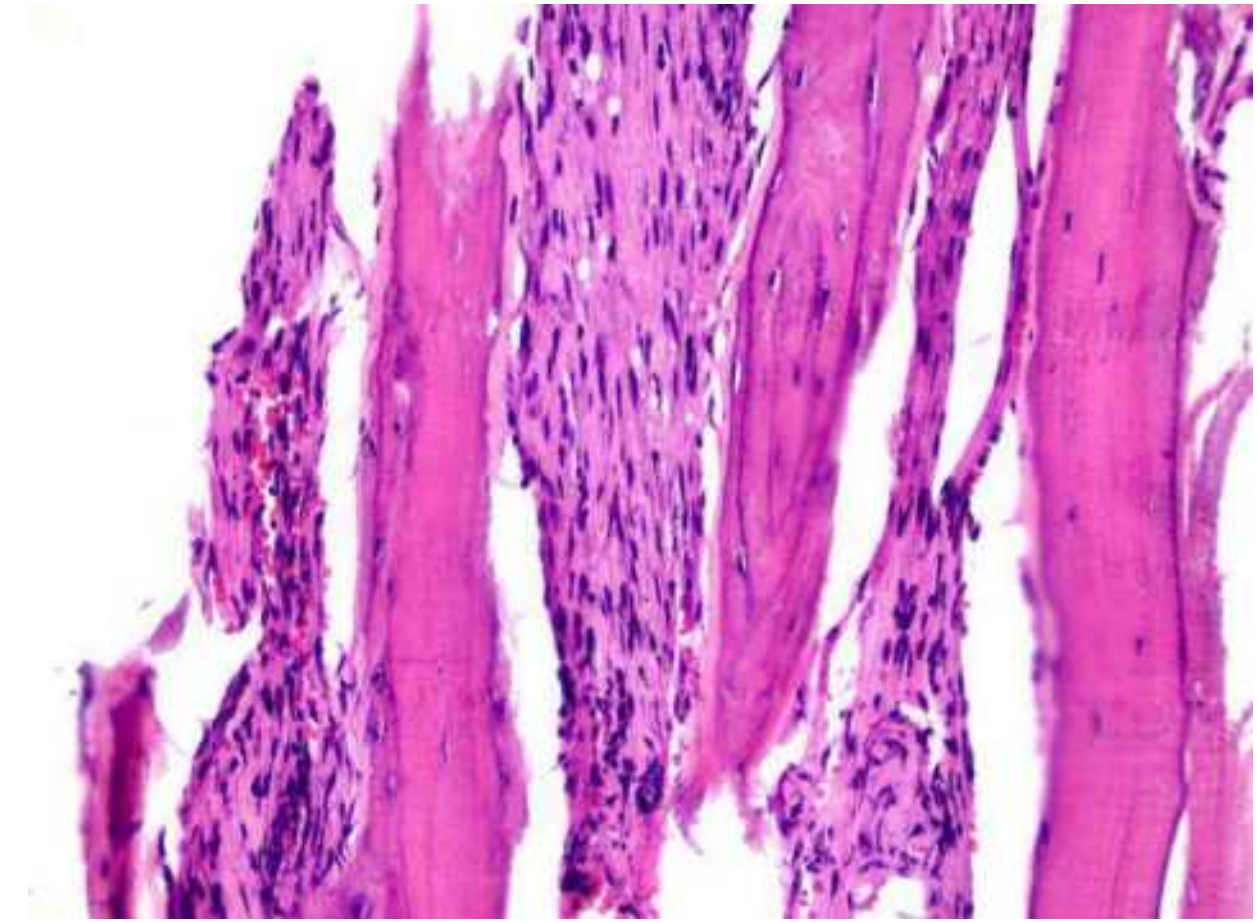
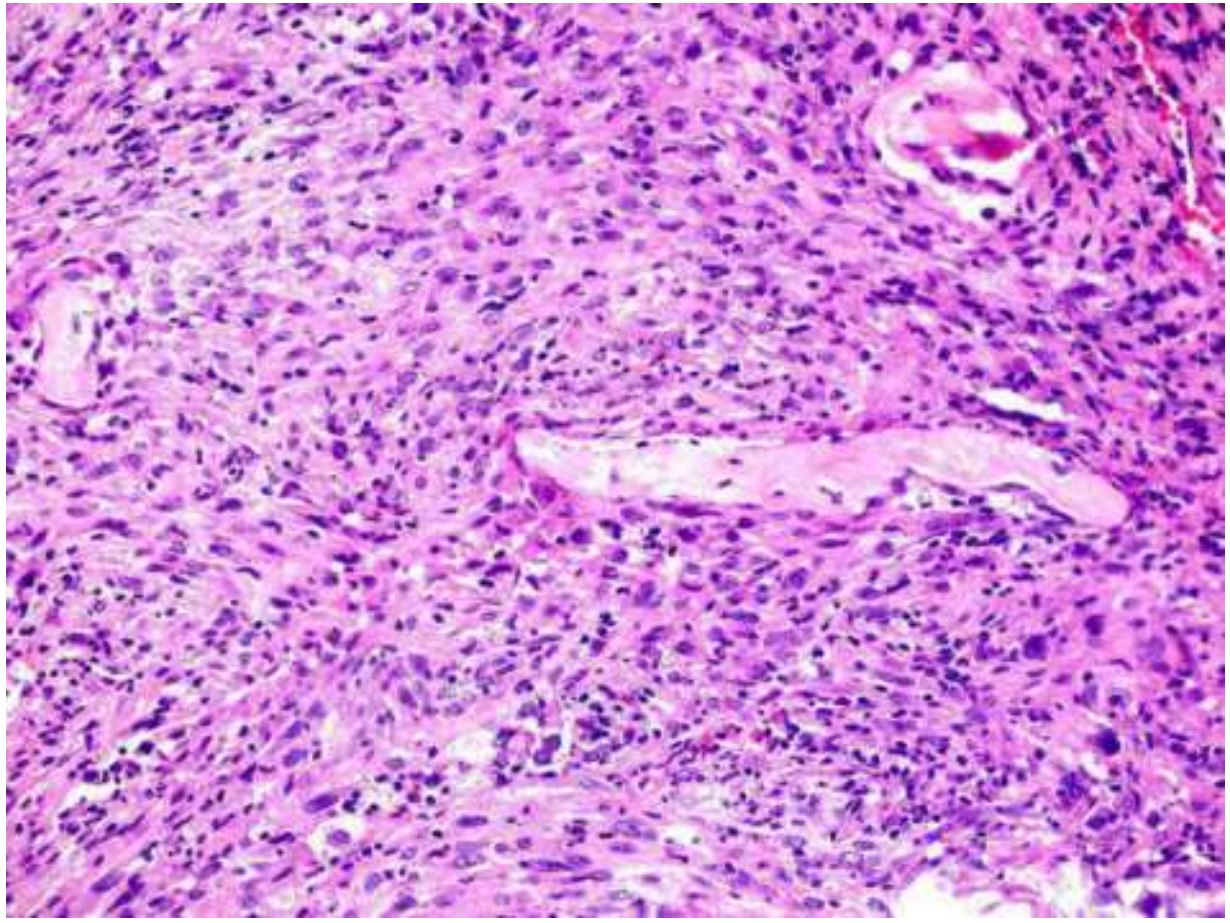
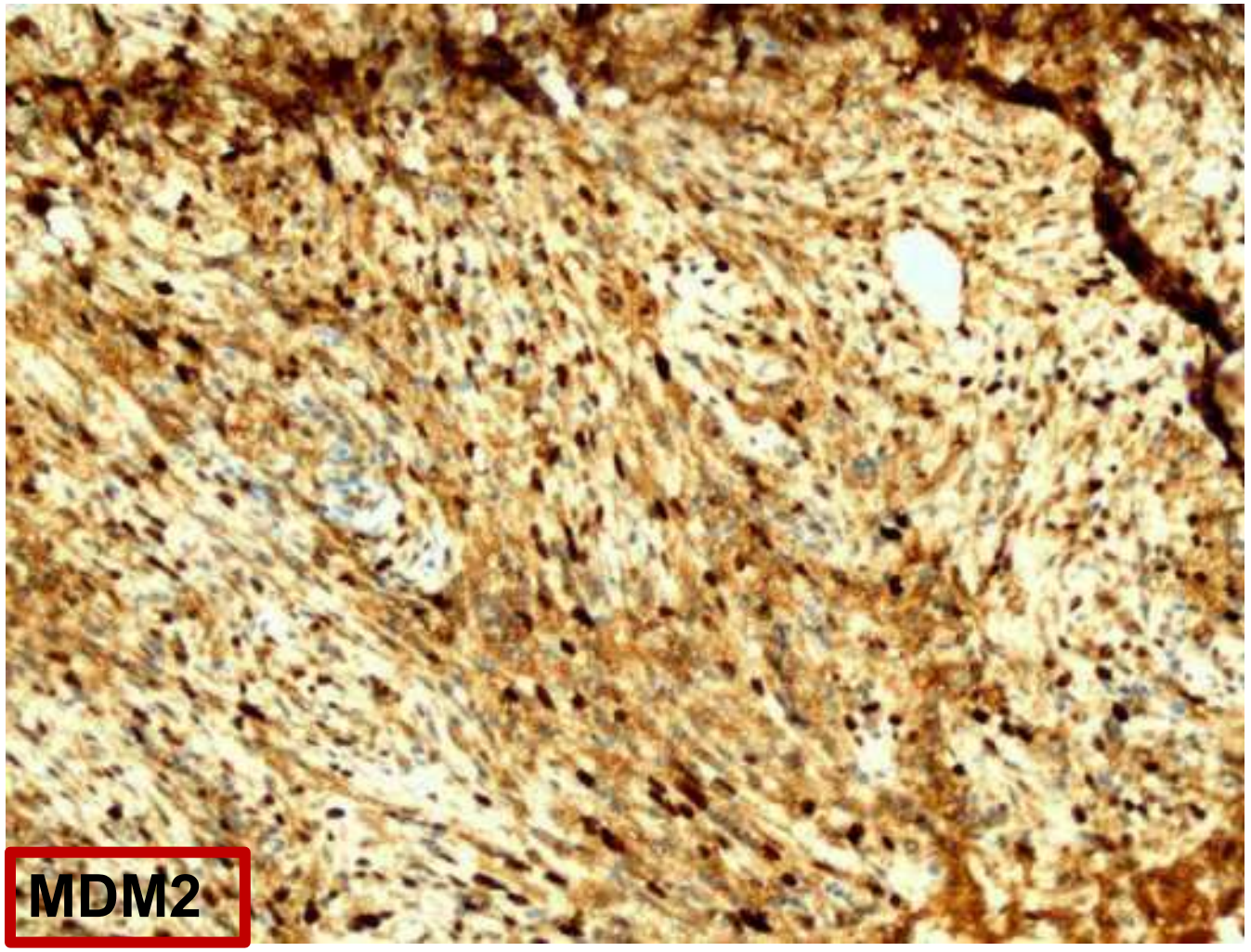


Image size: 768 x 768
View size: 1761 x 1376
A
3-747-914 (23 y , 23 y)
Tc - To rax Simple Alta Resolucio n
Toras Alta Resolucion -Thorax
Length: 4.714 mm
Length: 3.799 mm
Made in Horos







Diagnosis: Dedifferentiated Parosteal Osteosarcoma

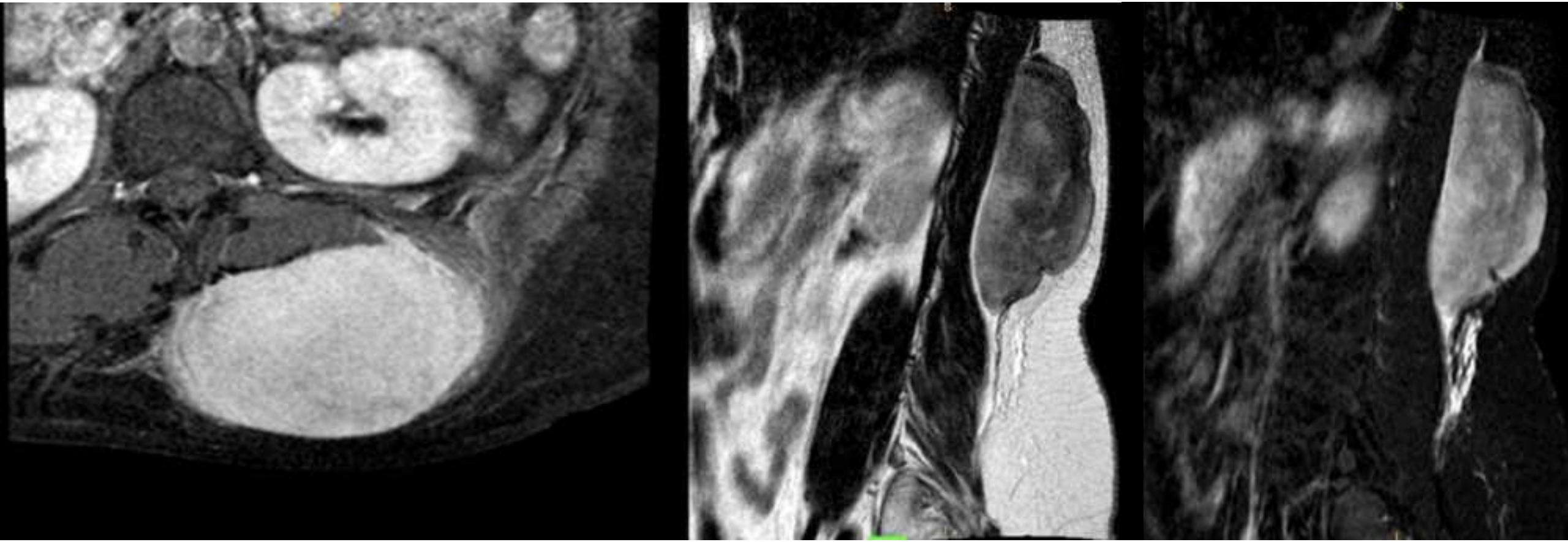
Questions

- Neoadjuvant chemotherapy + limb salvage surgery ?
- Limb salvage surgery + adjuvant chemotherapy?
- Limb salvage surgery + follow-up?
  - Chest CT Scan and local imaging studies at 2 months post-surgery

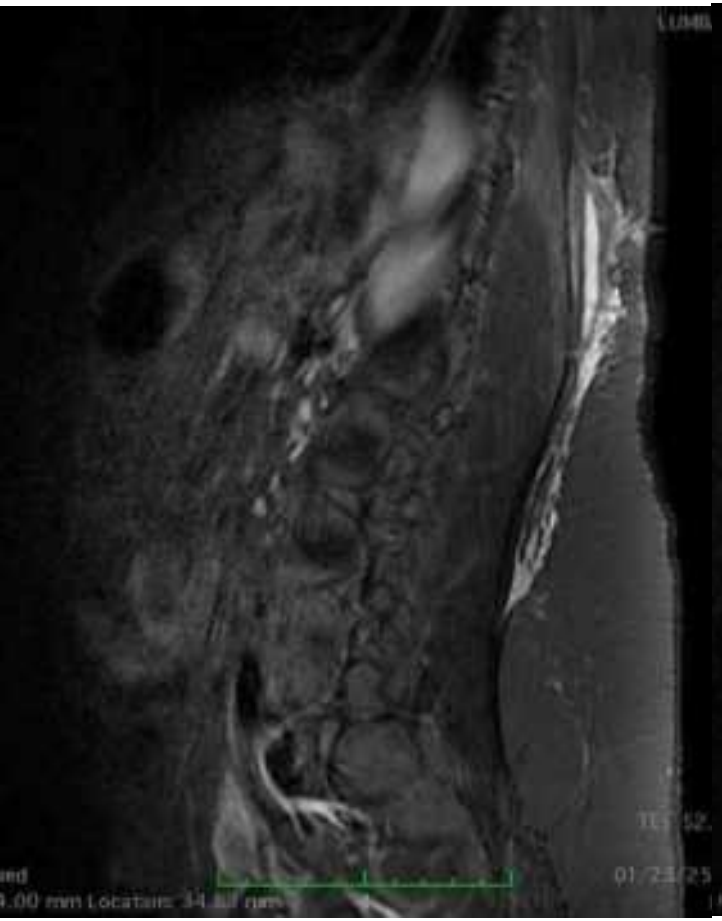
Patient #2 Panamá

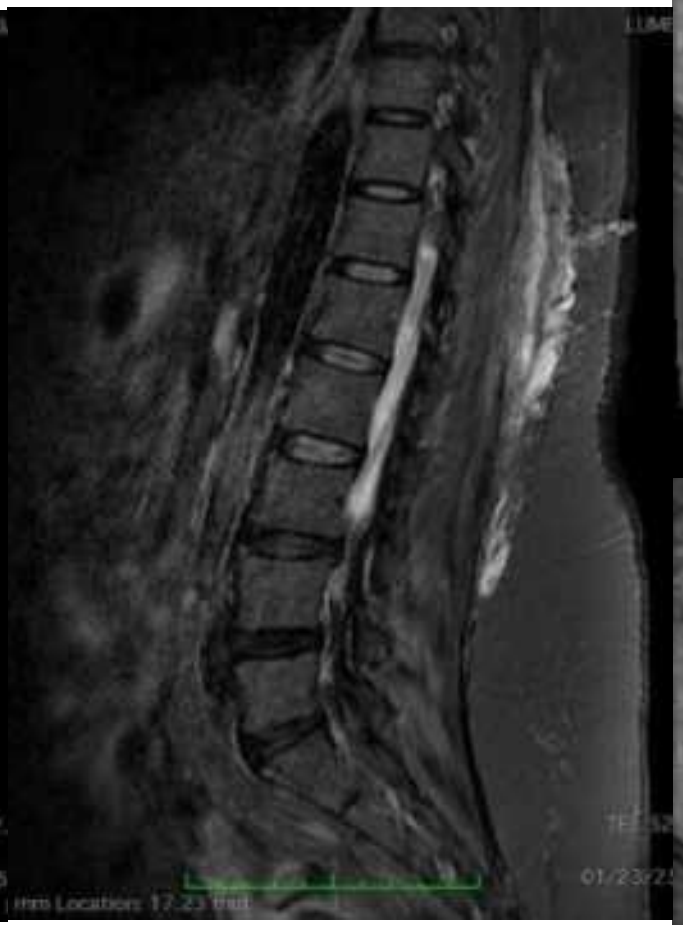
- 50-year-old female
- No prior history of disease; cholecystectomy (May 2023)
- One-year history of a painful mass in the lumbar region
- August 2024: Biopsy performed at another center (*benign spindle cell tumor)*
- November 2024: Wide-margin resection and flap reconstruction by oncology and plastic surgery teams.
- Persistent pain
- On physical exam
  - No palpable mass at the wound site. No pain on palpation

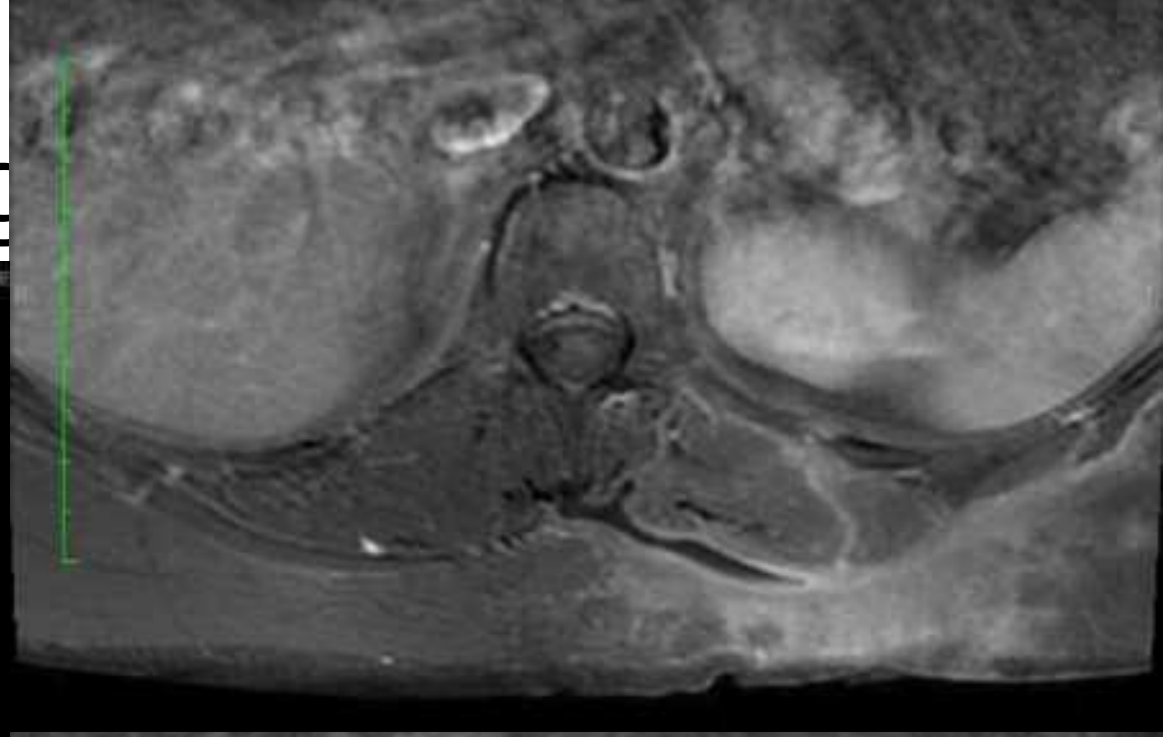
Imaging studies

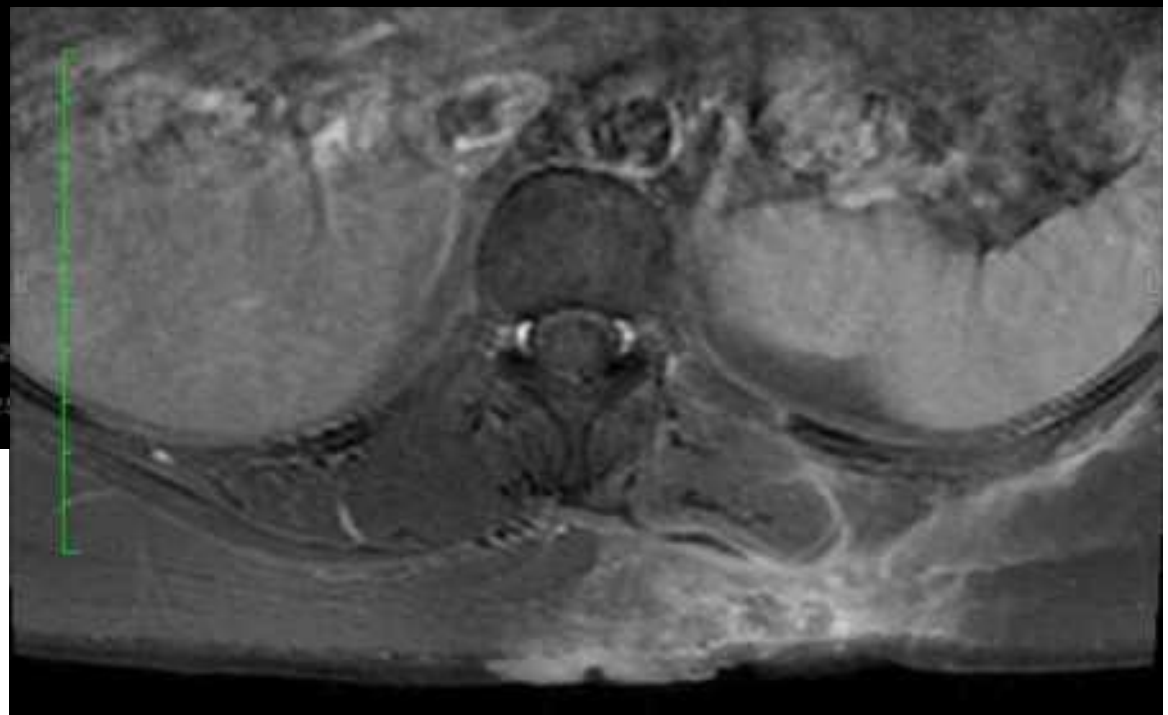


Post operative imaging

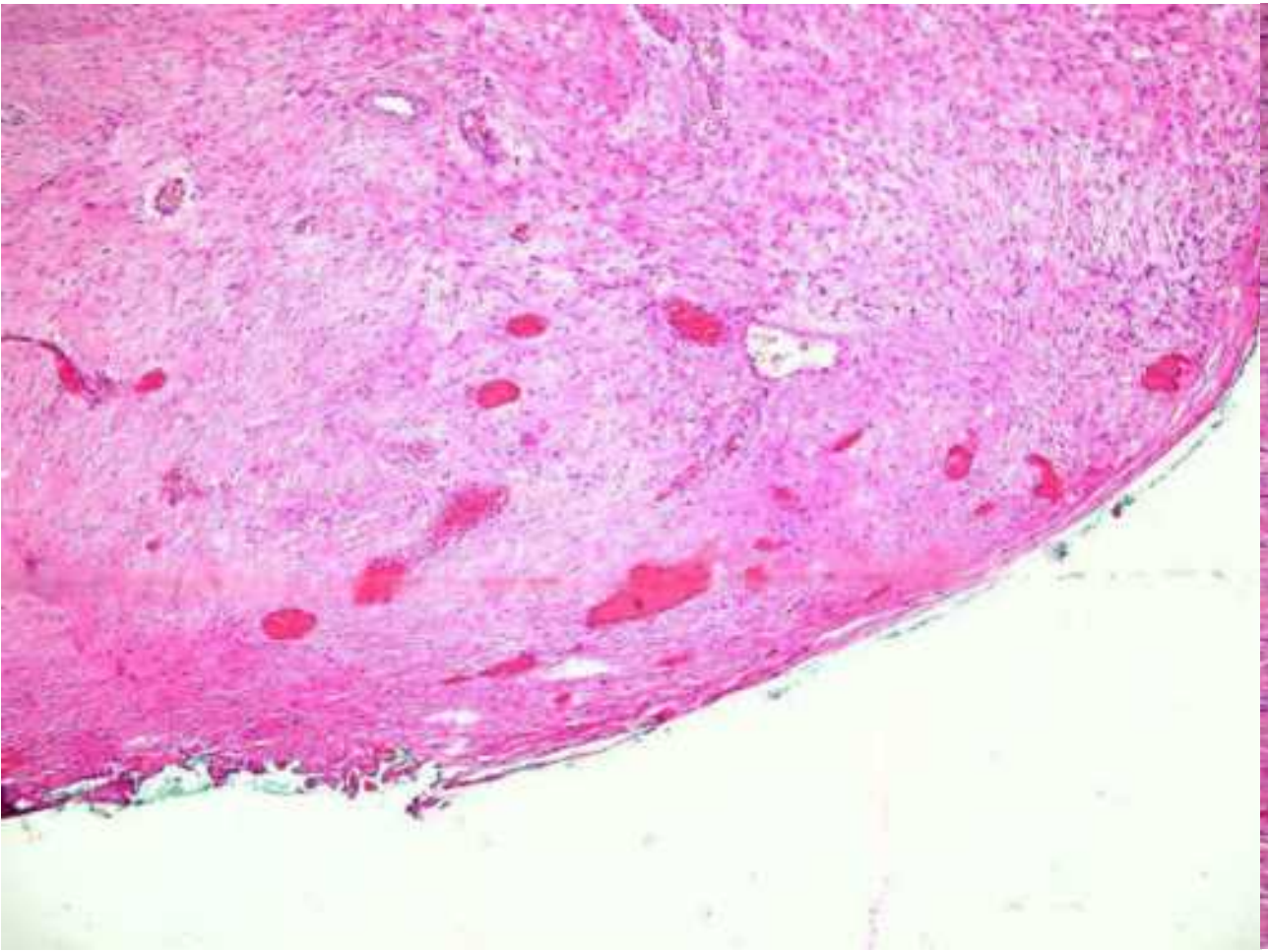


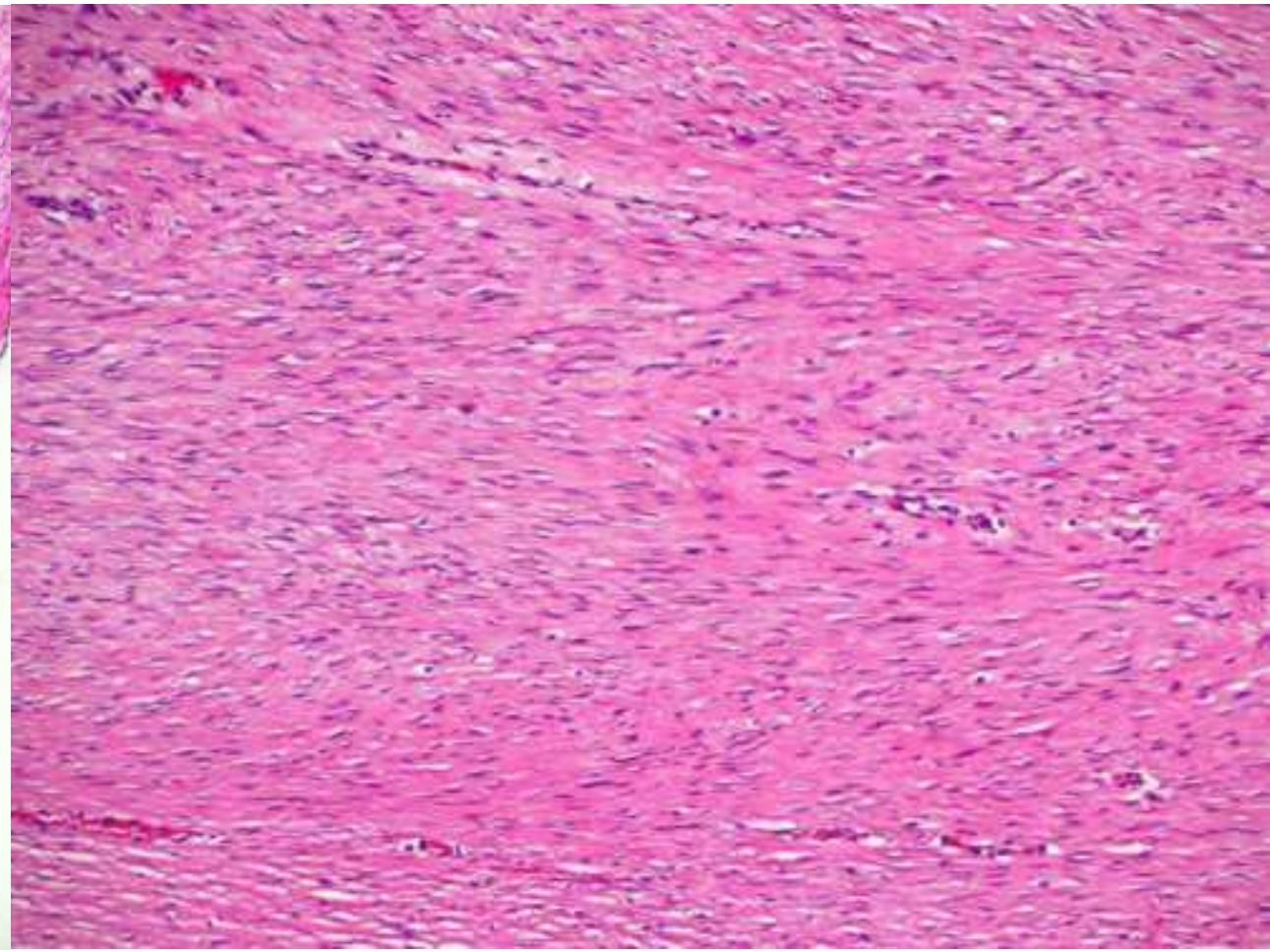


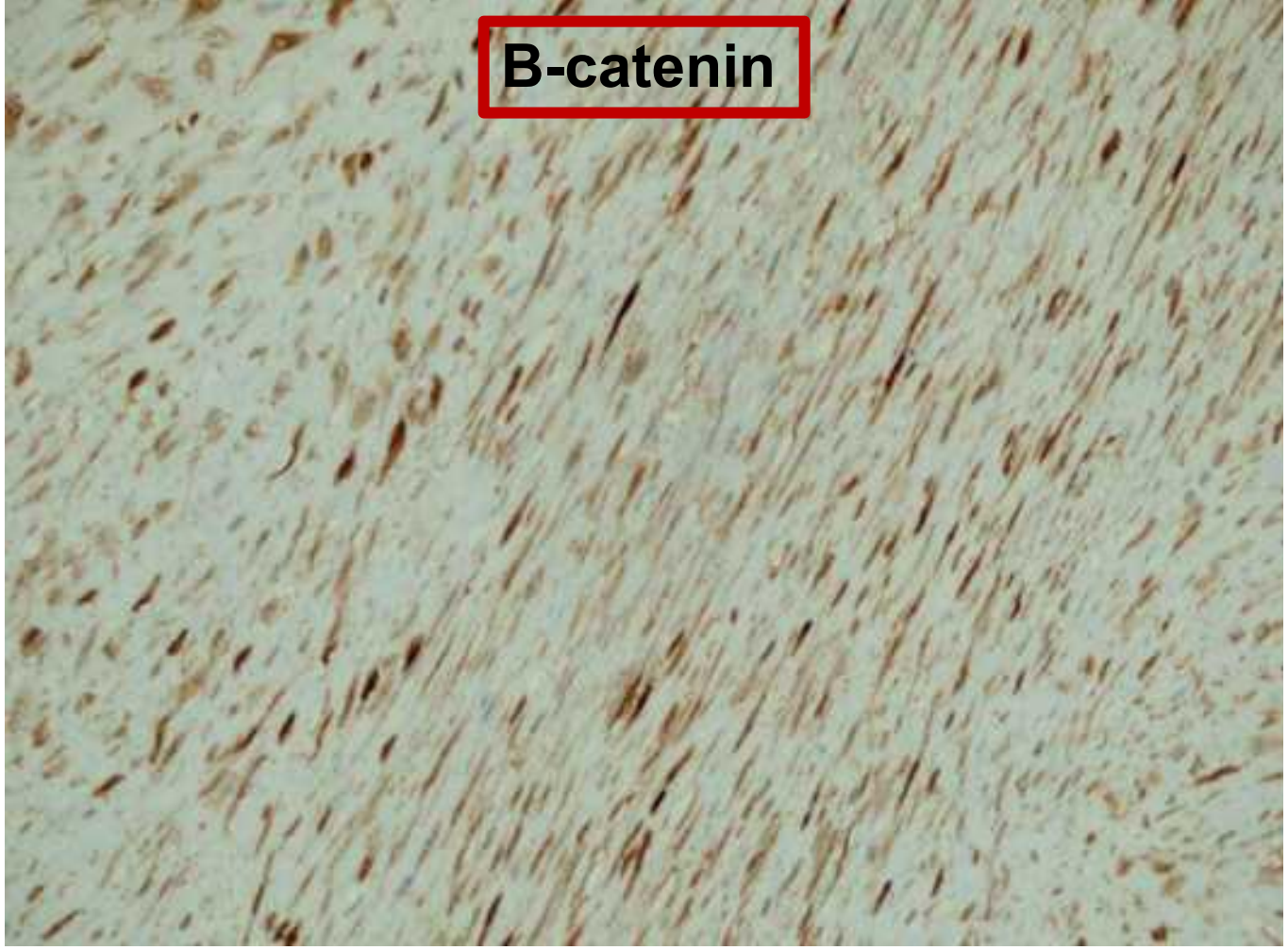




No macroscopical disease on imaging







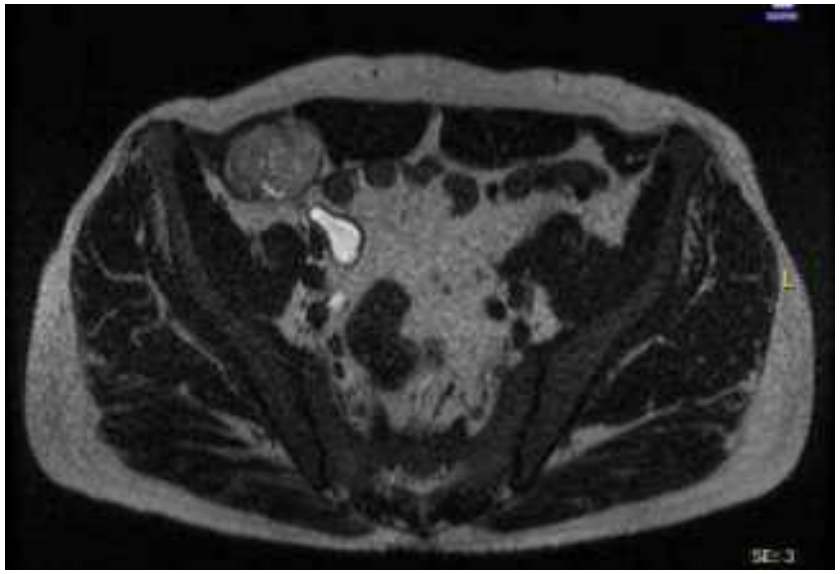
Diagnosis:
Desmoid fibromatosis
Margins: microscopically positive (distal and medial)

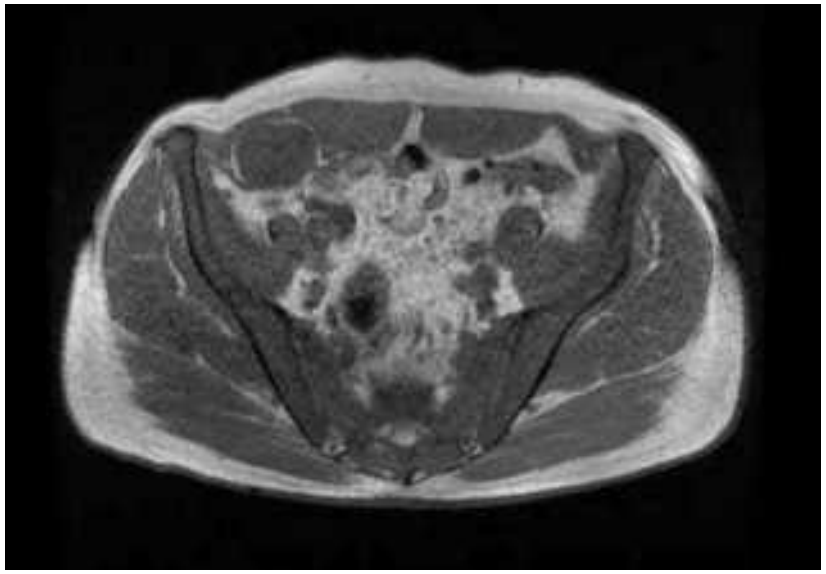
Question

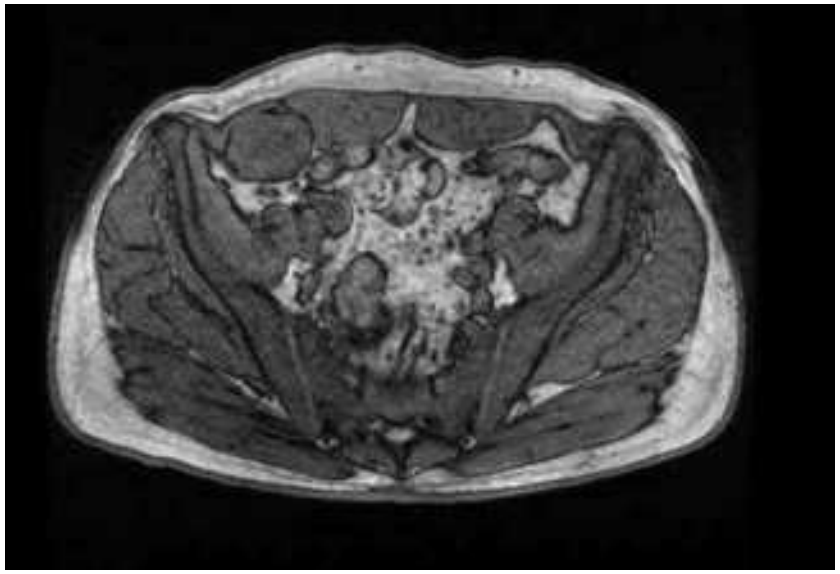
- What is the optimal follow-up strategy?
  - Frequency and preferred imaging modality for recurrence monitoring?
- If recurrence is diagnosed, what is the best treatment approach?
- What is the best second-line treatment option?
- Recommendations for pain management?

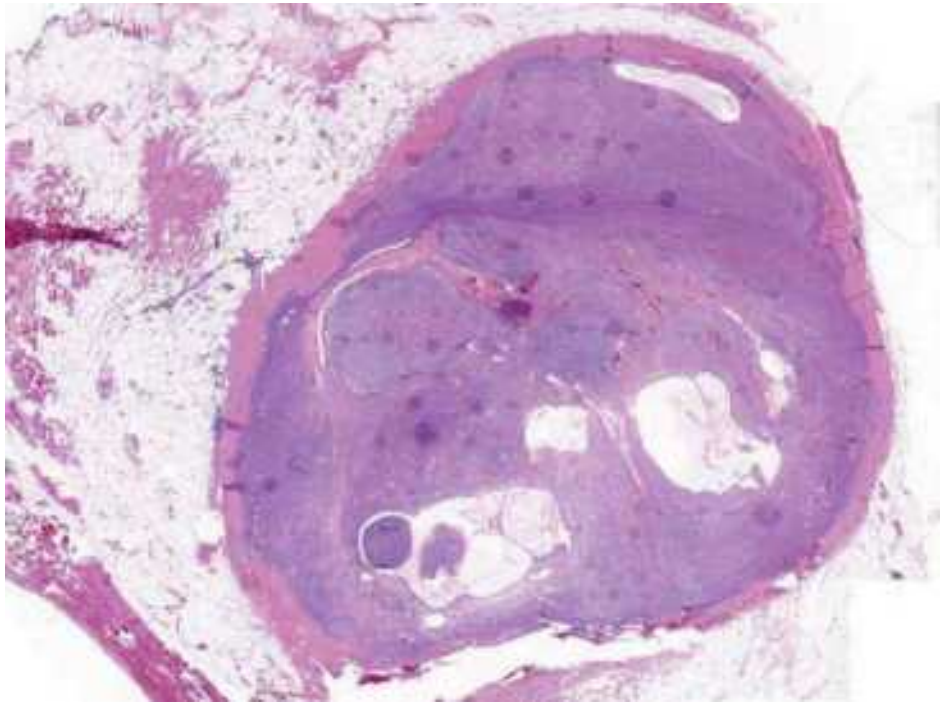
Patient #3 Valencia, Spain

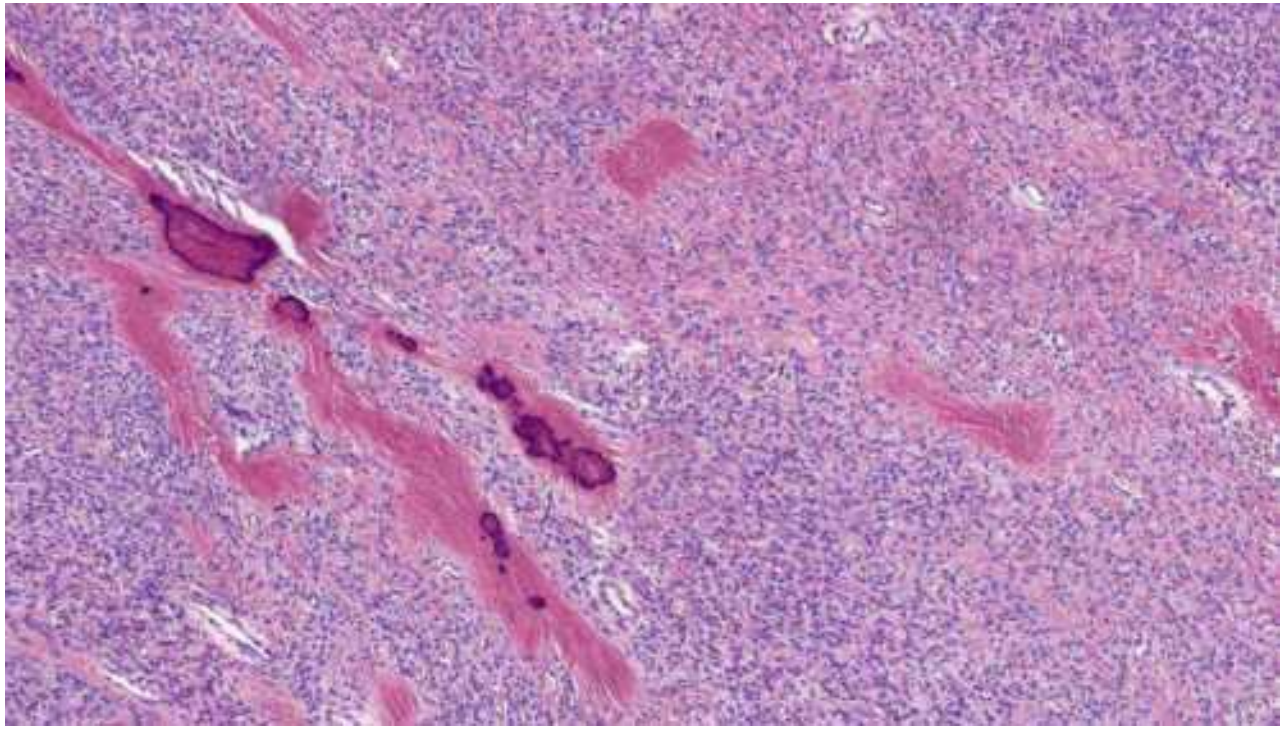
- 75-year-old male with a soft tissue tumor in the abdominal wall



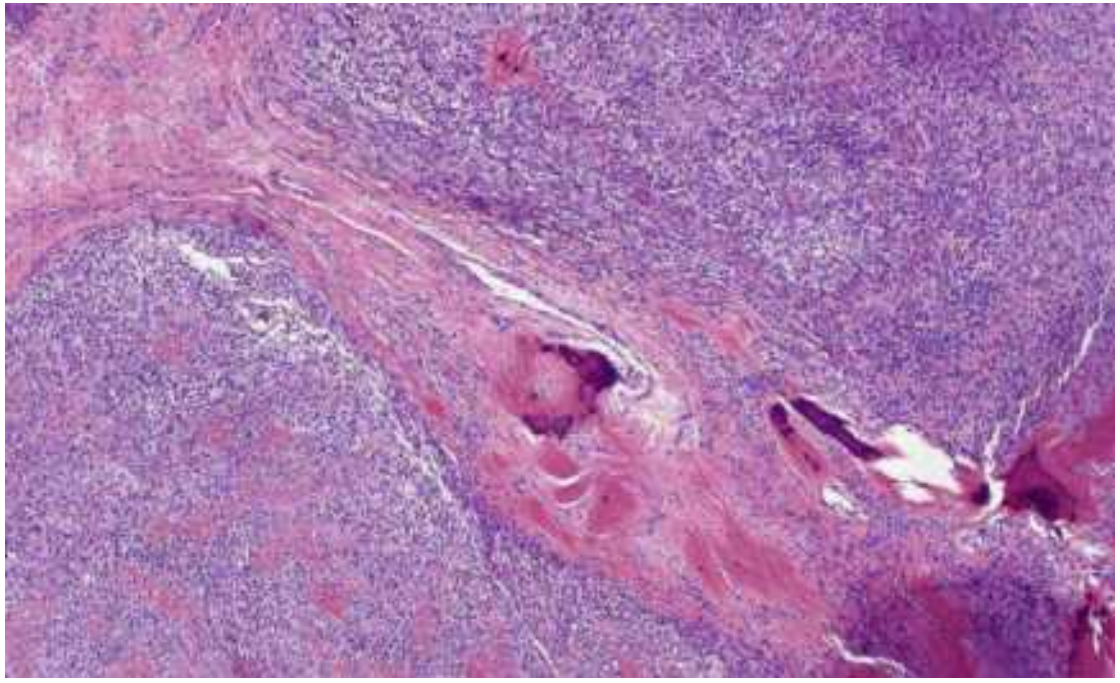


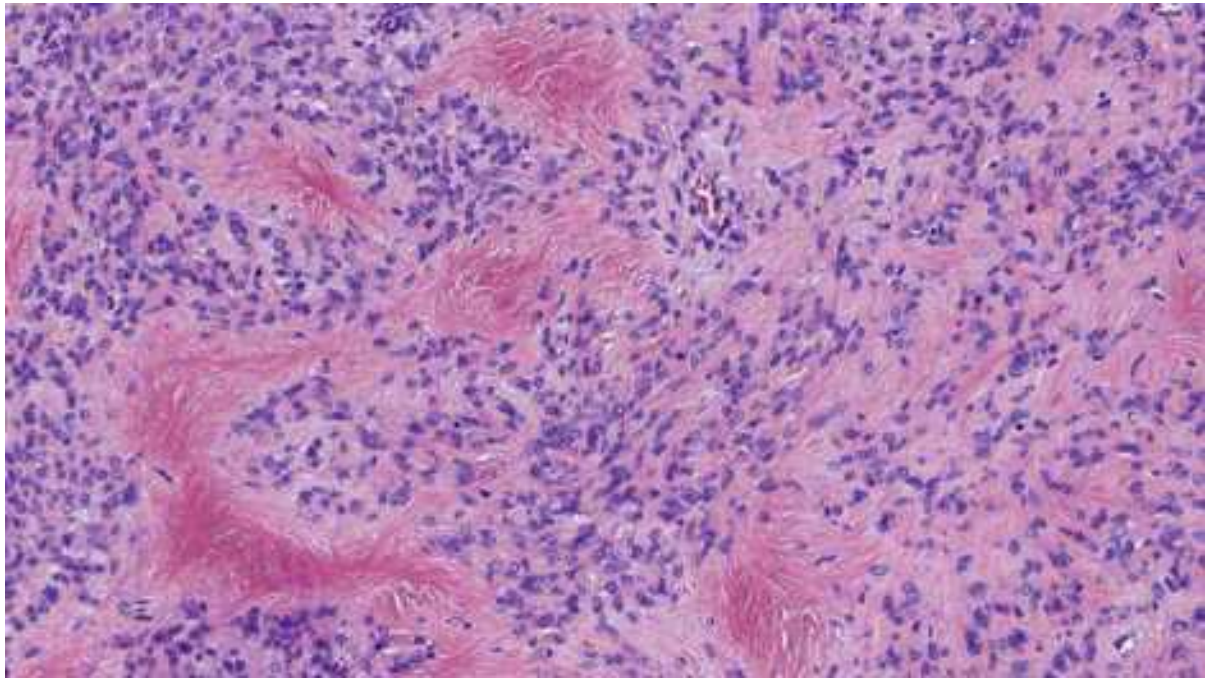


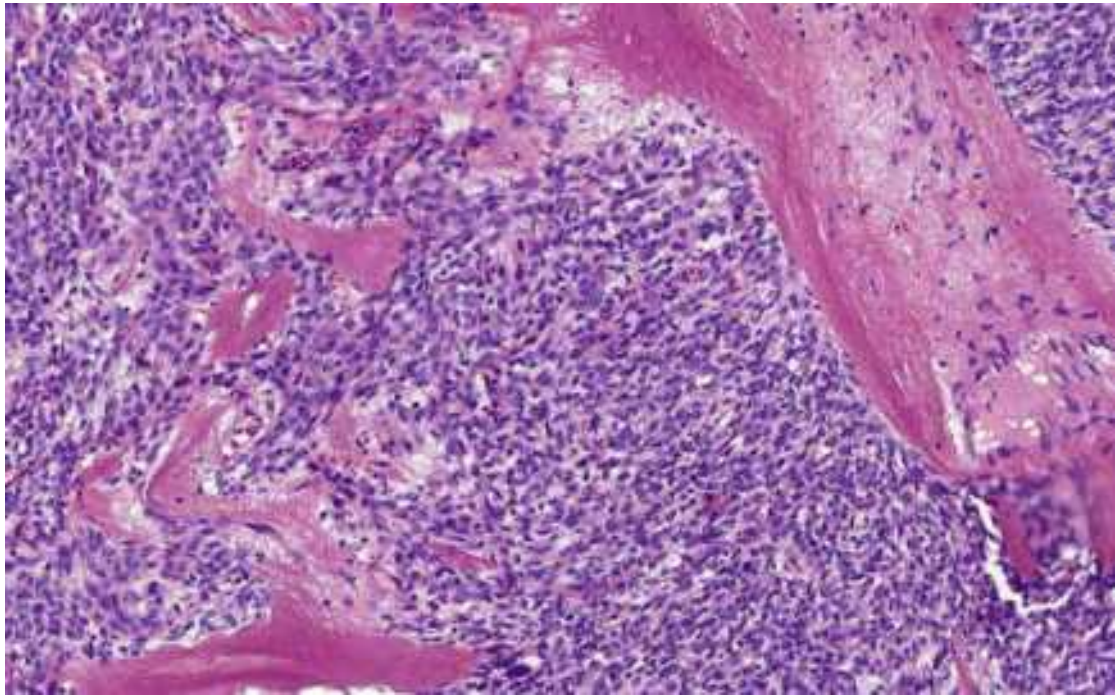


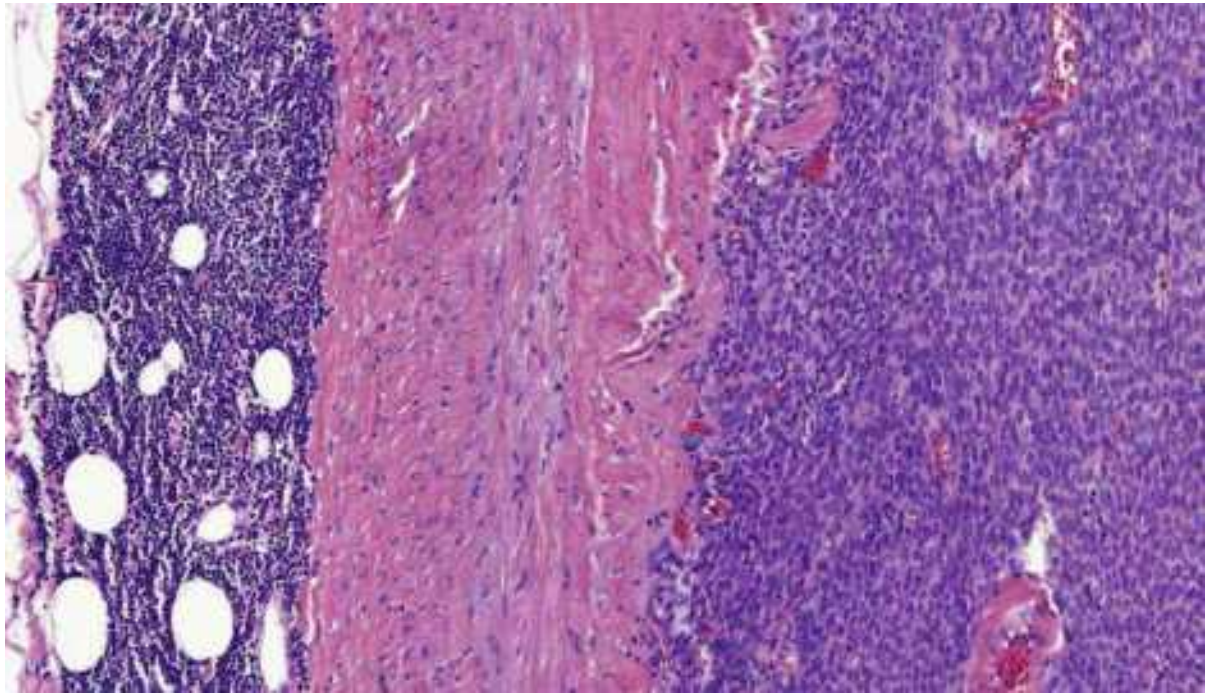


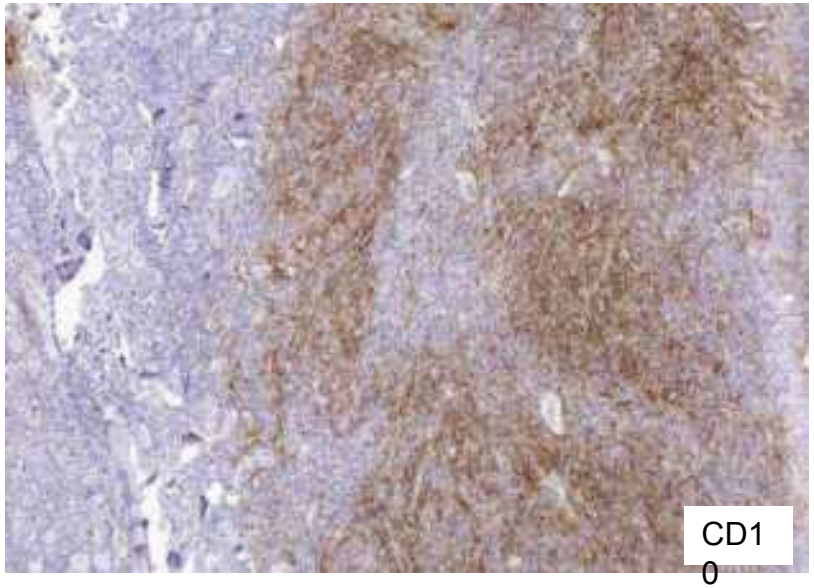
SELNET Instituto Valenciano de Oncología (February 27): Isidro Machado, Reyes Claramunt,

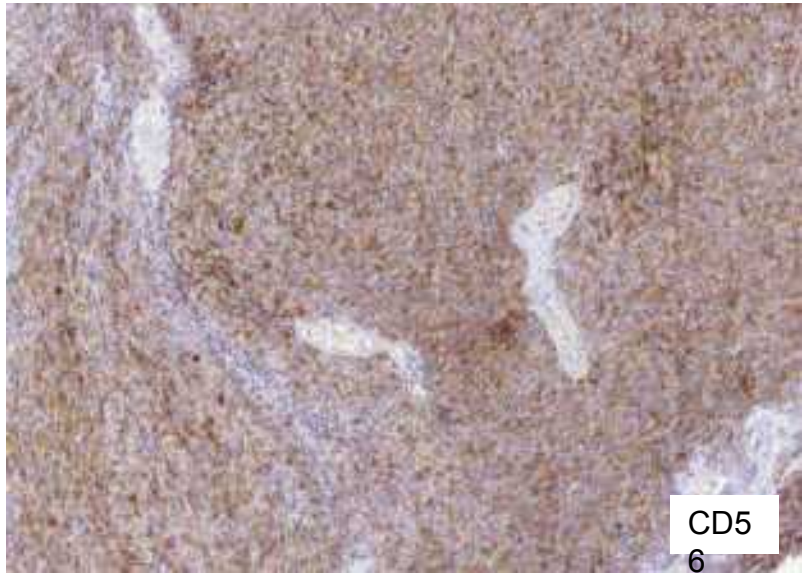


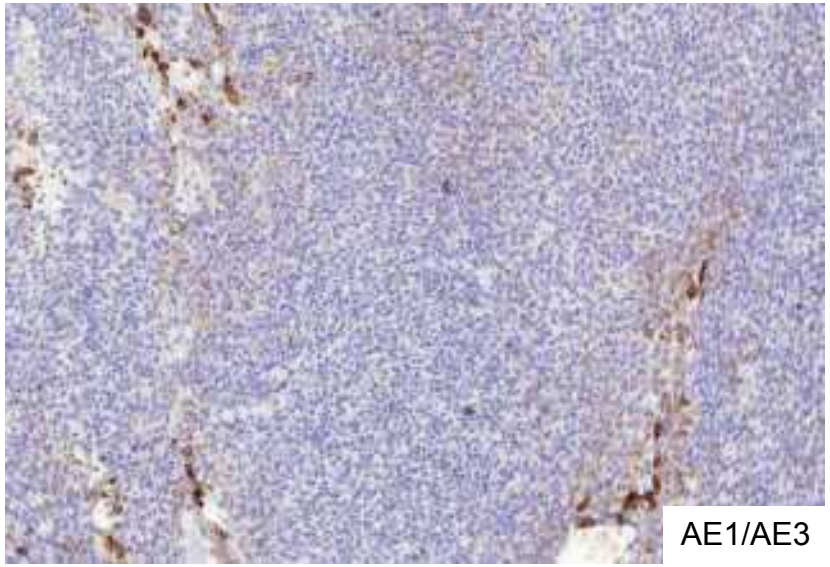


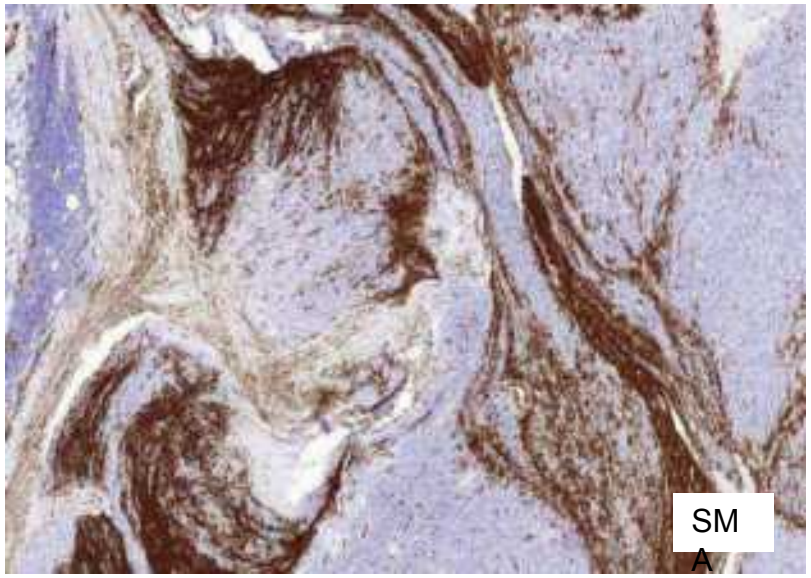


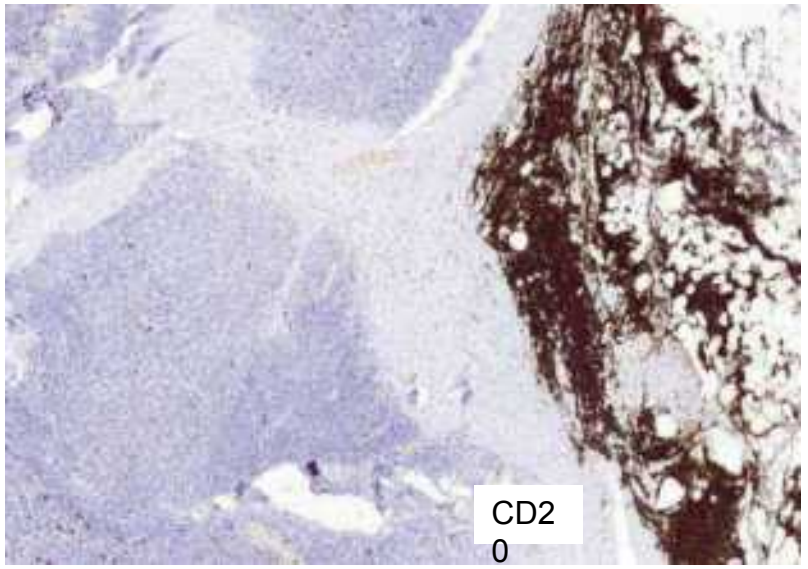


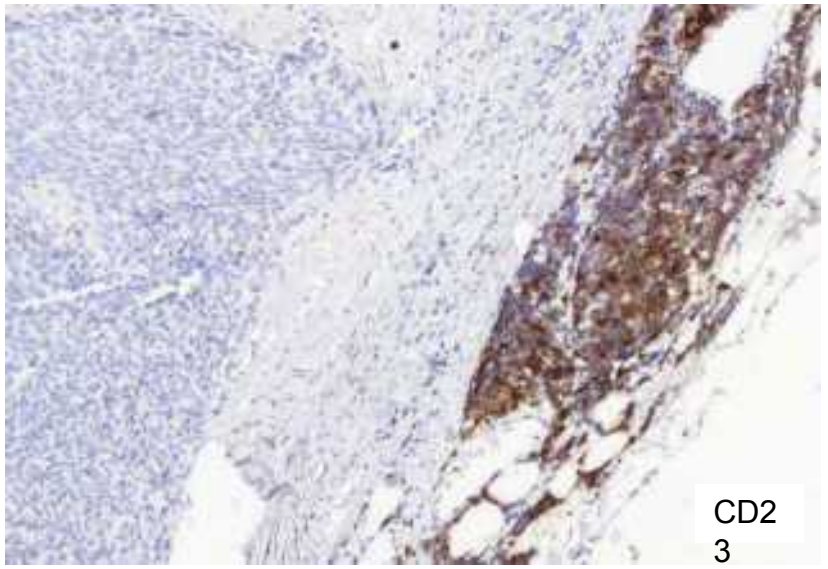

CD1
0


CD5
6

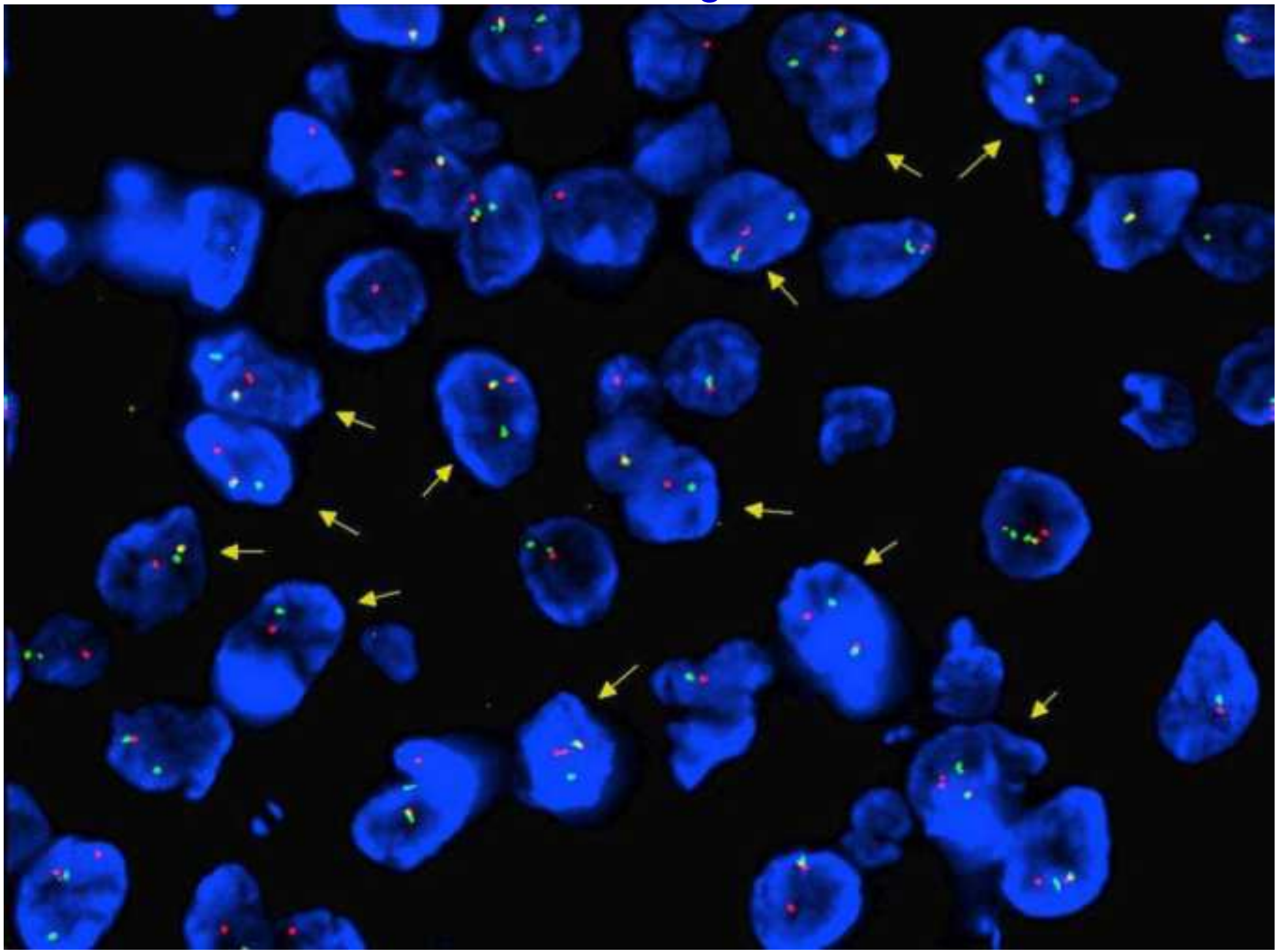

AE1/AE3


SM
A


CD2
0


CD2
3

PHF1 rearrangement



Courtesy Prof. Gregory Charville. Stanford. USA

Ossifying fibromyxoid tumor in association with chronic lymphocytic leukemia

- EWSR1::PATZ1 sarcoma
- Soft tissue myoepithelial tumor
- Low-grade fibromyxoid sarcoma/sclerosing epithelioid fibrosarcoma
- Extraskeletal myxoid chondrosarcoma
- Myxoid leiomyosarcoma

Patient #4 Omán

- 60-year-old female

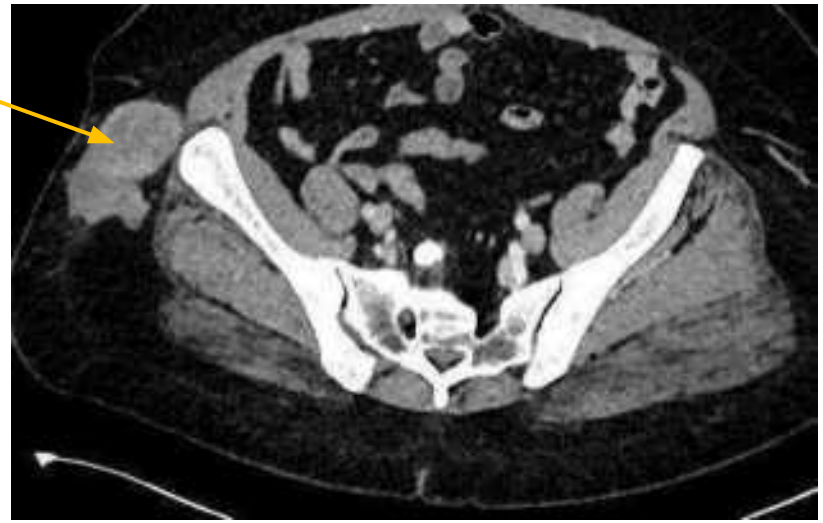
Comorbidity and PMH

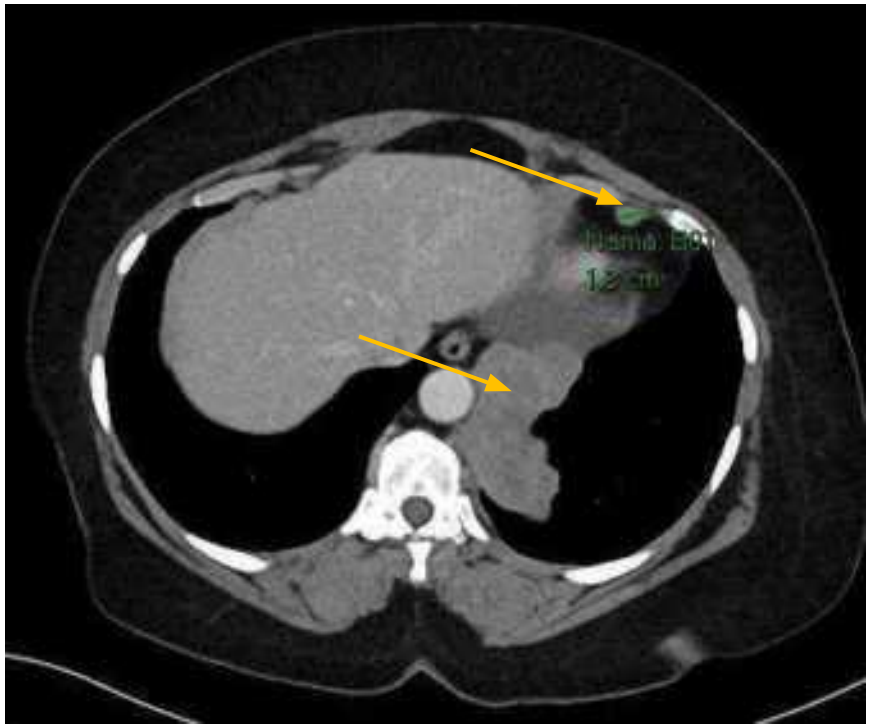
Asthma – Seretide
HTN –
DLP
Obesity

She found a soft tissue mass in her abdominal wall 1 year ago, near asymptomatic. ECOG-0

CT September 2024

- Well defined lobulated pleural based inhomogeneously hypo-enhancing solid mass in left lower lung lobe.
  - Large well defined inhomogeneously enhancing lobulated mass lesion in subcutaneous tissue in right lateral gluteal region.
  - Few small pleural based nodular lesions in bilateral chest

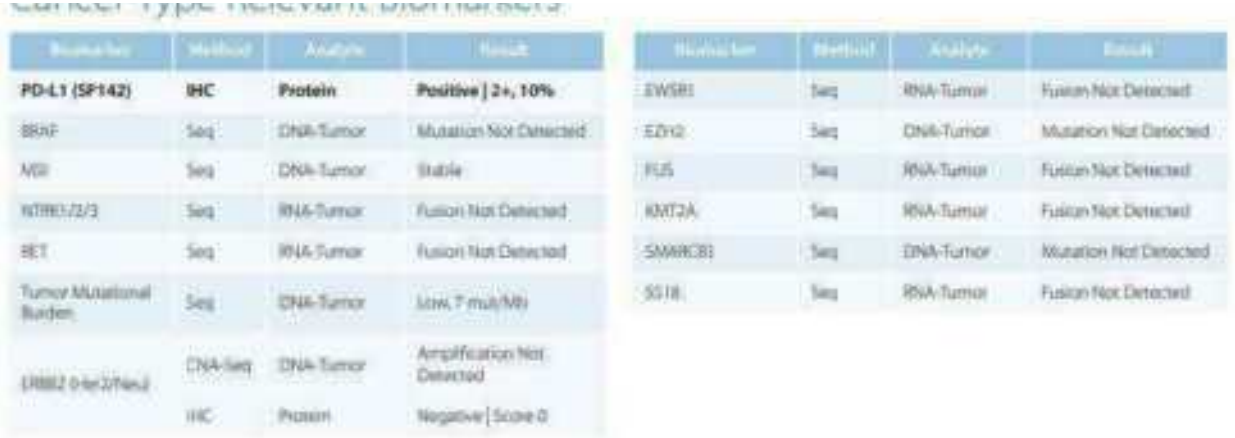




Lung biopsy: Intermediate grade sarcoma associated with dense mixed inflammation. It was

Soft tissue biopsy sent to reference pathology lab:

The biopsy shows an epithelioid and spindle cell neoplasm with myxoid-to-edematous stroma and admixed cells with enlarged, atypical nuclei. There are scattered multinucleated tumor cells, and there is readily identified mitotic activity. These appearances are non-distinctive, and unfortunately immunohistochemistry is not helpful in determining a line of differentiation. We found focal expression of CDK4, focal weak expression of MDM, and diffuse expression of HMGA2, while stains for S-100, SOX10, CD34, EMA, pan-K, SMA, desmin, LCA, PU.1 and GLI1 are entirely negative. Because of our findings, we thought it worthwhile to repeat FISH testing, and I learned from the lab that our FISH is negative for *MDM2* amplification. In the end, I can do no better than to render the descriptive diagnosis of an **unclassified epithelioid and spindle cell sarcoma, low-grade** in this biopsy specimen. I have reviewed this case with Dr. Jason Hornick, who concurs. Given the reported distribution of disease including multiple abdominal wall masses and a pleural mass, it seems distinctly possible that this



PD-L1 (SP142)	**IHC**	**Protein**	**Positive | 2+, 10%**
BRAF	Seq	DNA-Tumor	Mutation Not Detected
MDI	Seq	DNA-Tumor	Stable
NTRK1/2/3	Seq	RNA-Tumor	Fusion Not Detected
RET	Seq	RNA-Tumor	Fusion Not Detected
Tumor Mutational Burden	Seq	DNA-Tumor	Low, 7 mut/Mb
ERBB2 (Her2/Neu)	CNA-Seq	DNA-Tumor	Amplification Not Detected
	IHC	Protein	Negative | Score 0

EWSR1	Seq	RNA-Tumor	Fusion Not Detected
EZH2	Seq	DNA-Tumor	Mutation Not Detected
FUS	Seq	RNA-Tumor	Fusion Not Detected
KMT2A	Seq	RNA-Tumor	Fusion Not Detected
SMARCB1	Seq	DNA-Tumor	Mutation Not Detected
SS18	Seq	RNA-Tumor	Fusion Not Detected

- Thoracotomy: pleural infiltration with multiple small nodules

New HPE:

- intermediate grade malignant sarcoma.
- pleomorphic spindle tumor cells show diffuse strong positivity for desmin and Fli-1 with a proportion of these showing strong to moderate positive staining for myo-D1and very few positive for myogenin. They are negative for caldesmon, SMA, muscle specific actin, SOX-10, s-100, MDM-2, EMA, CK- AE1/AE3 and pan-CK, ALK-1, calretinin, STAT-6, USP-6, D2-40, CD-34, ERG, Factor VIII and Histone 3. They show non-specific cytoplasmic staining for CD-99. Background staining noted for CD-31, and USP-6 are noted. INI-1 expression is retained
- The morphology and IHC is most consistent with a rhabdomyosarcoma arising on a background of a histiocyte rich ‘inflammatory rhabdomyoblastic tumor’.

- We started on Dox Ifo Mesna

In summary:

60 y o patient with advanced RMS on the background of inflammatory Myofibroblastic Tumor

Questions to the board:

What is the best medical treatment?

What Is the primary tumor site: lung or soft tissue?

Any role of PD-L1?