症例1

名古屋大学

31yo Man
Right axillary neurofibroma

Medical history

- Diagnosed with NF1 in infancy
- Oct, Year X: Referred to our department
- Mar, Year X+1: Whole-body MRI demonstrated multiple neurofibromas
- May, Year X+3: Rapid growth of the brachial plexus tumor (2 cm → 6 cm)
- Dec, Year X+3: Resection of the brachial plexus tumor (①)
- May, Year X+5: Recurrence of the brachial plexus tumor (②)
- Oct, Year X+5: Progressive enlargement of the tumor was observed
  Biopsy followed by tumor resection (③)

Past medical history: Unremarkable

Pathological findings

① Initial tumor resection

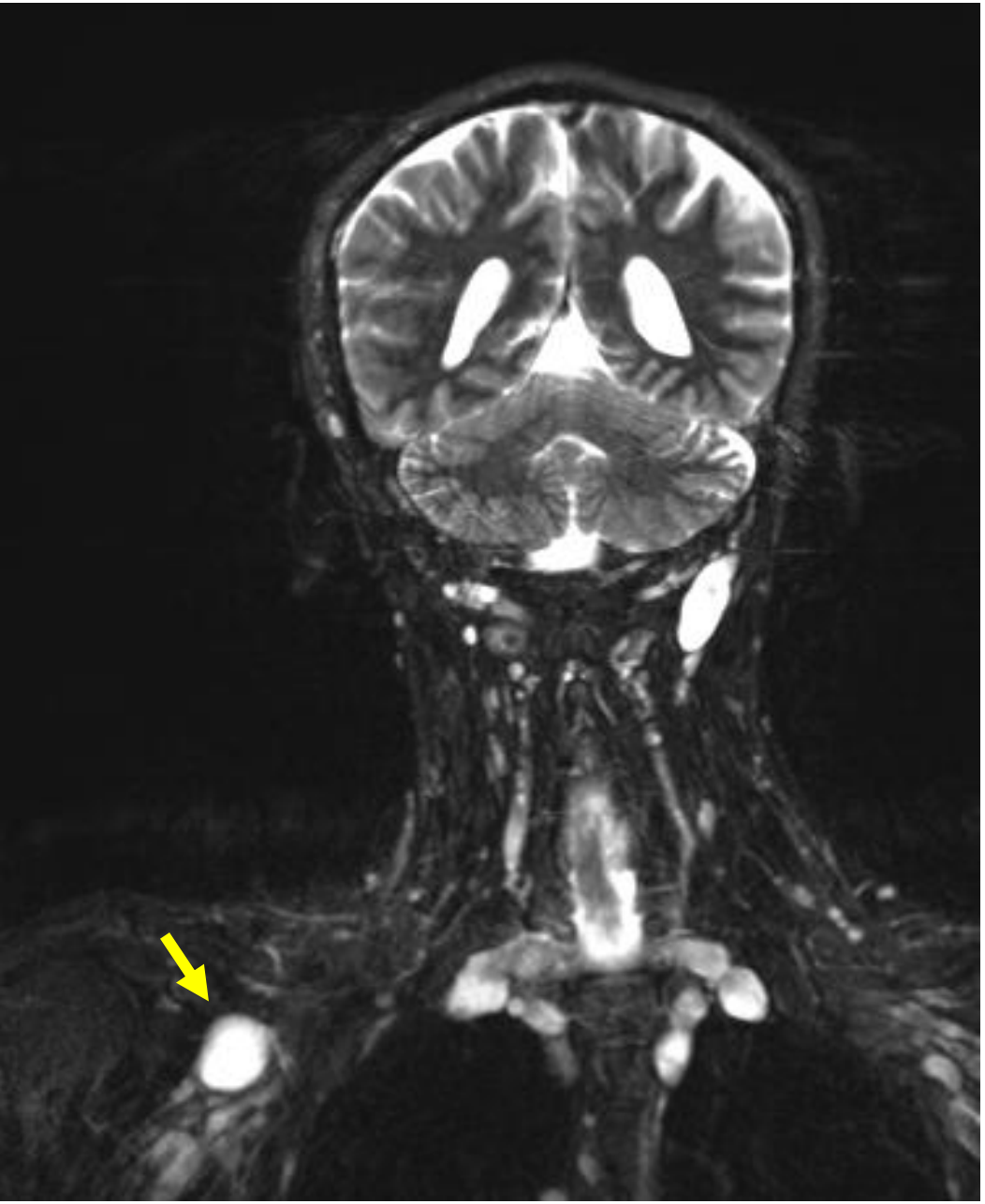
- Neurofibromatosis, soft tissue, resection
- S100(+), SOX10(+), H3K27me3(retain)

② Biopsy of a recurrent tumor

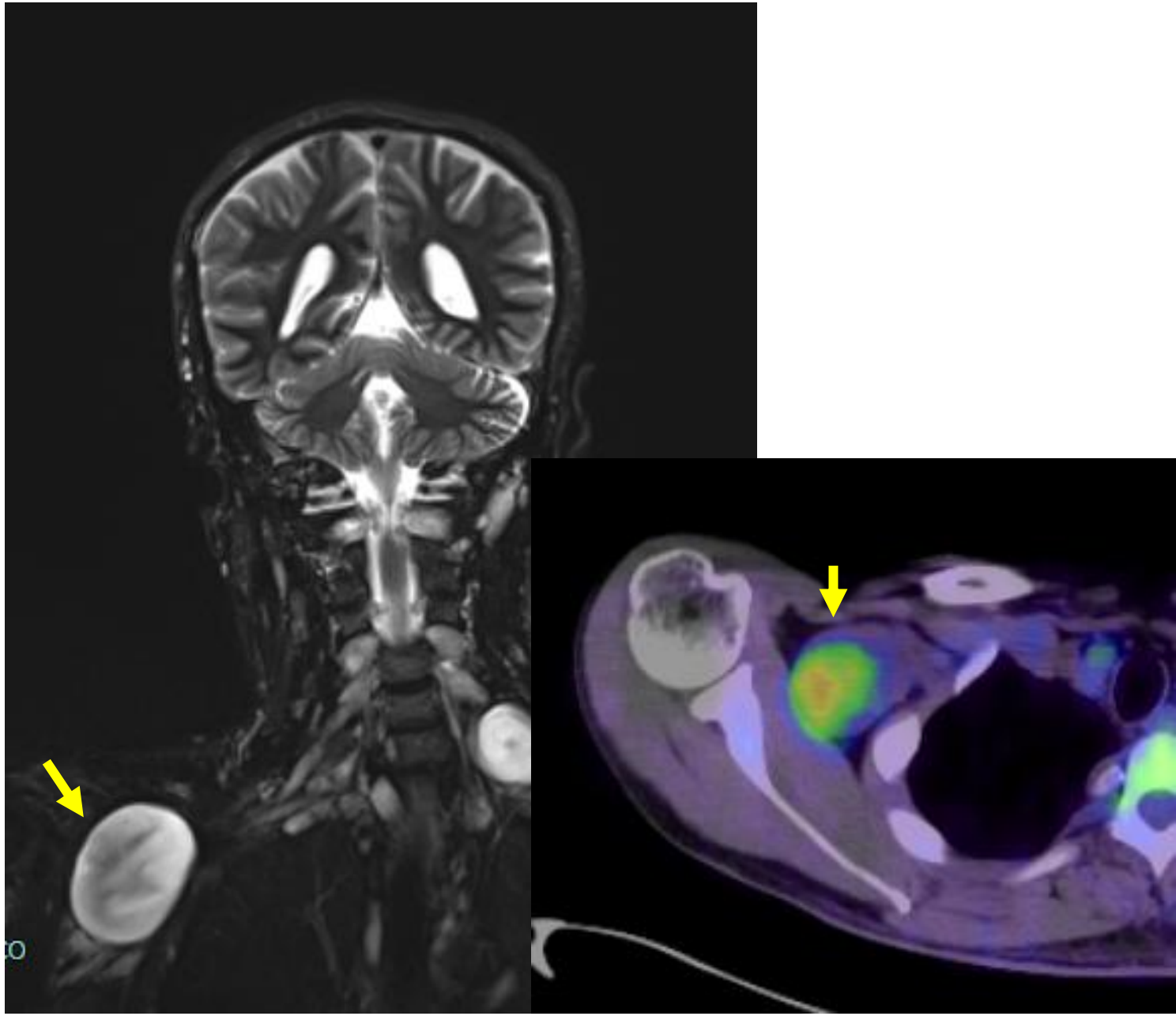
- Malignant peripheral nerve sheath tumor (MPNST), low grade, cellular neurofibroma or more
- S-100(-), SOX10(-), H3K27me3(loss), Ki-67;10%

③ Biopsy of recurrent tumor (rapid growth)

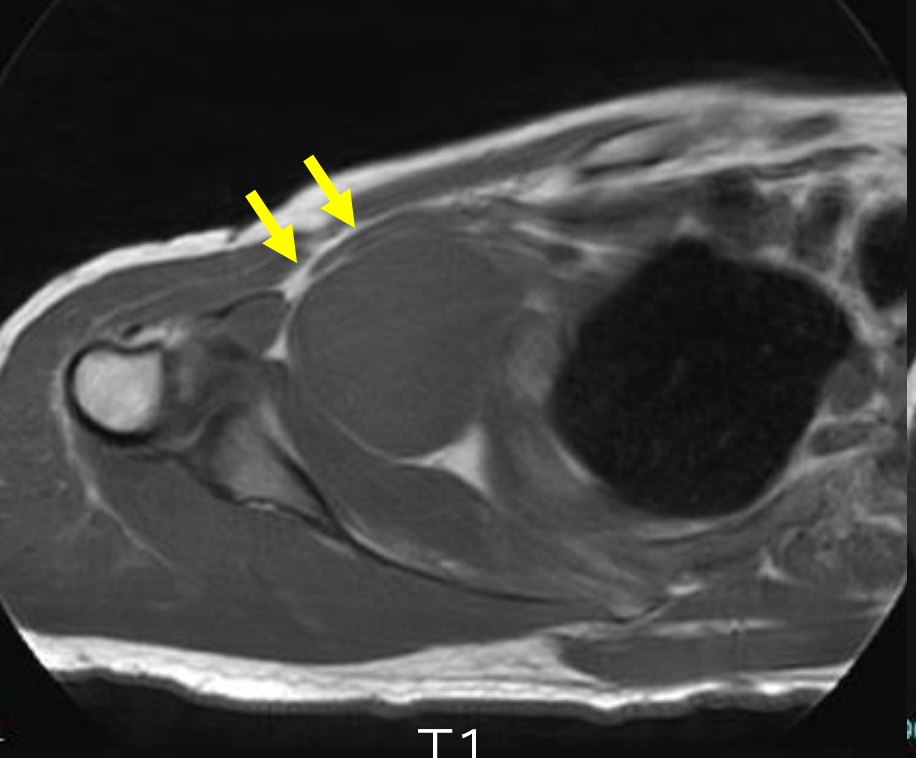
- Malignant peripheral nerve sheath tumor(MPNST), high grade
- S-100(-), SOX10(-), H3K27me3(loss), Ki-67;60%

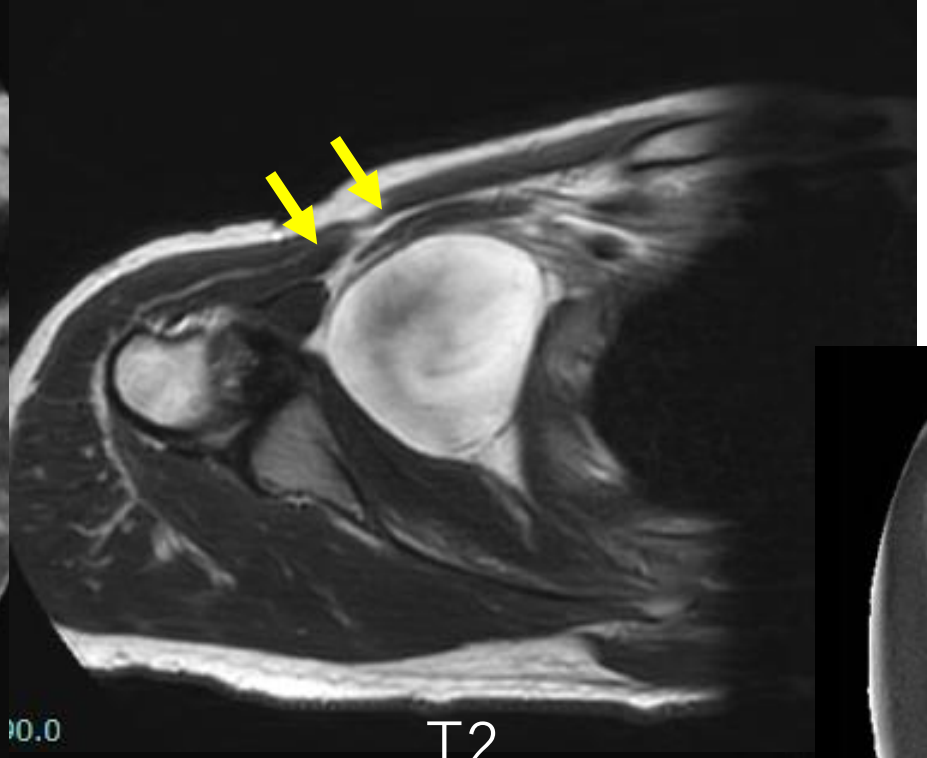


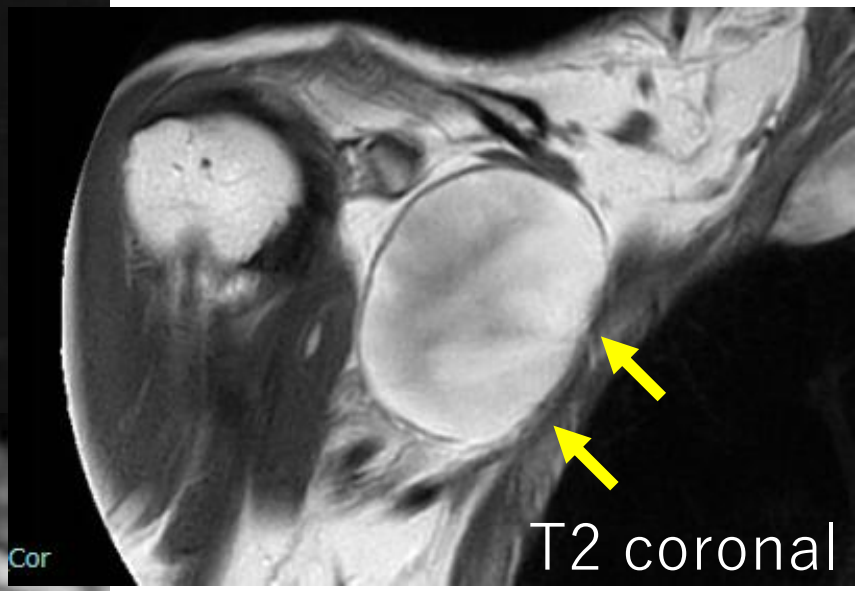
Mar, Year X+1

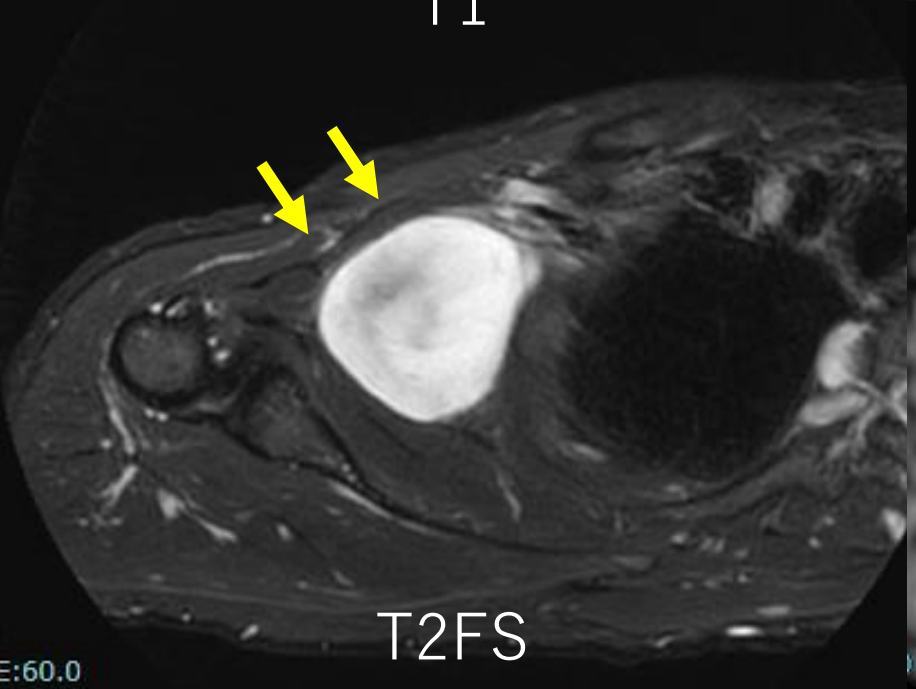


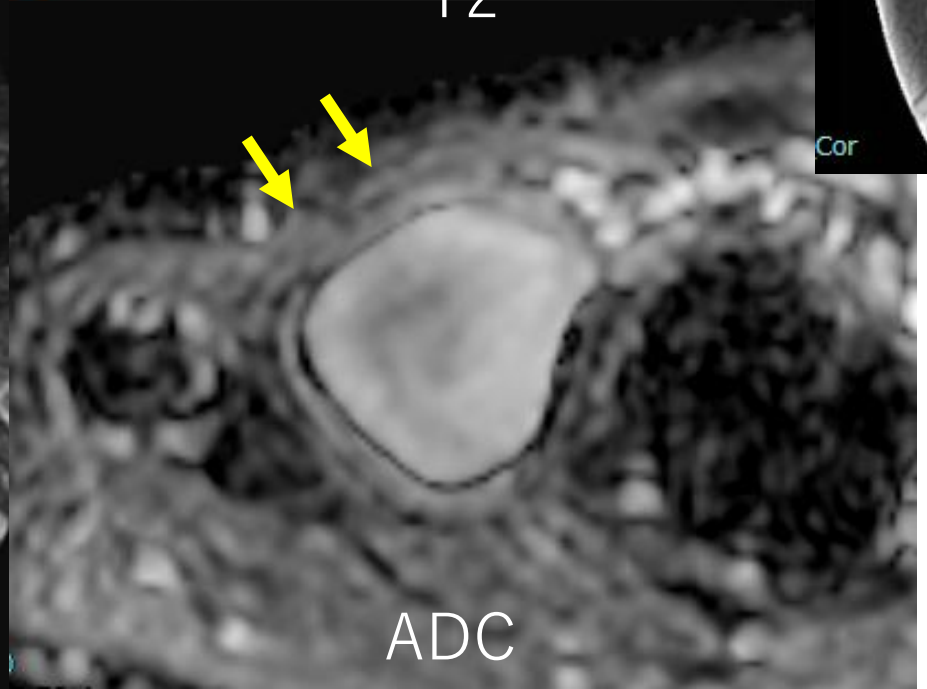
May, Year X+3



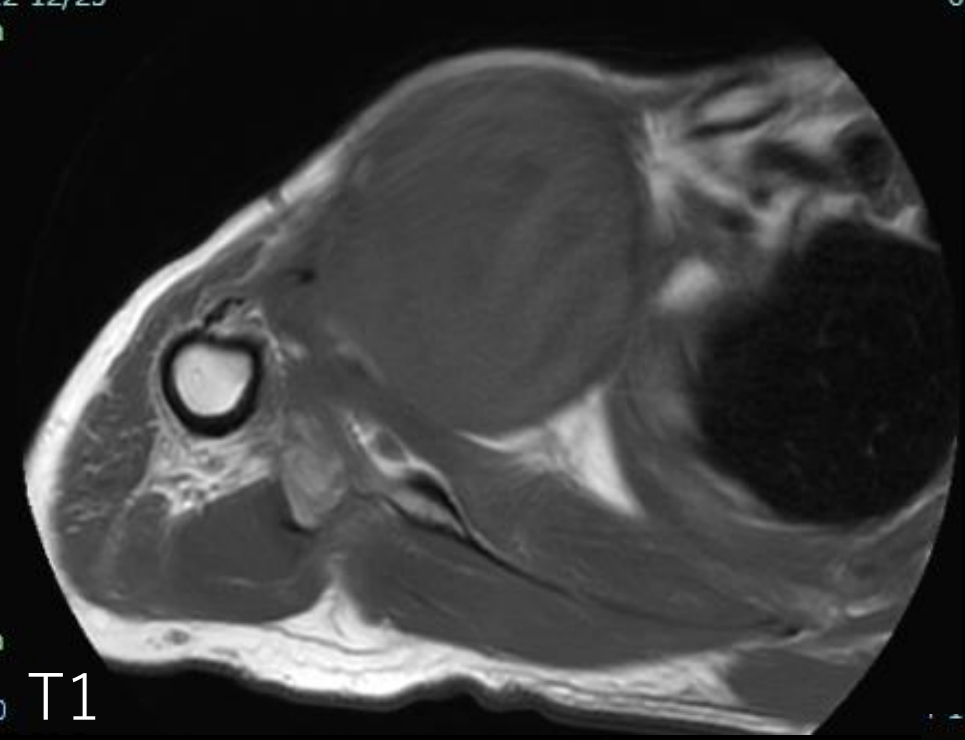


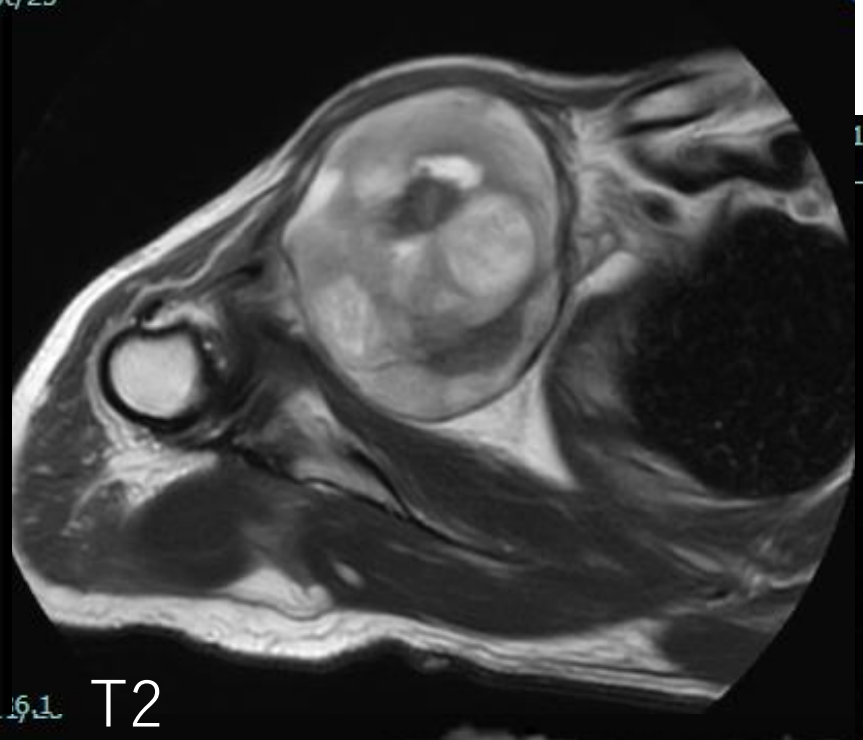


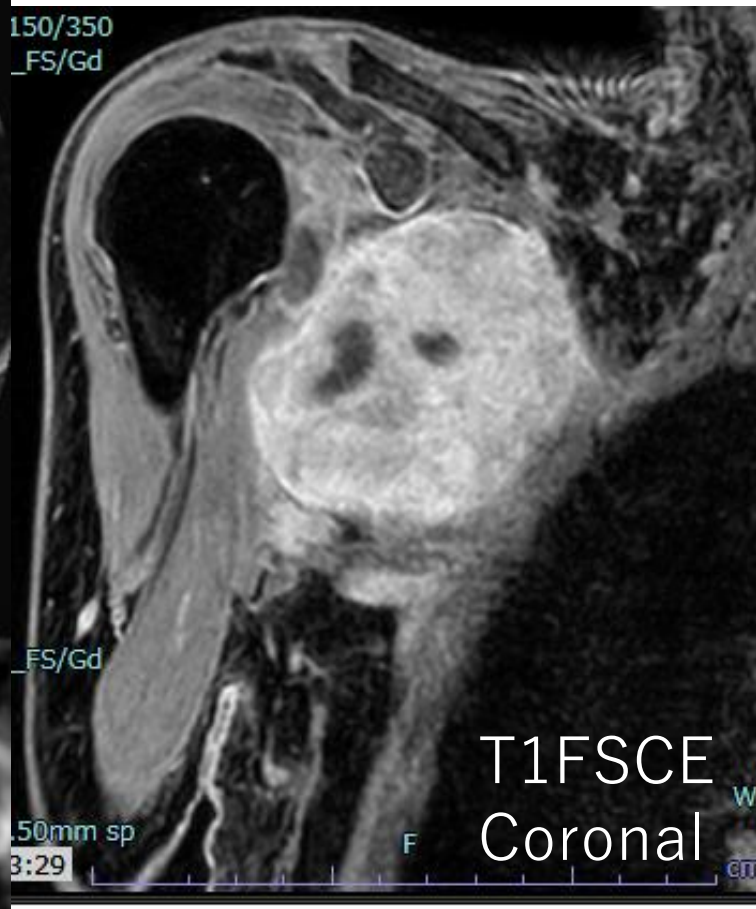


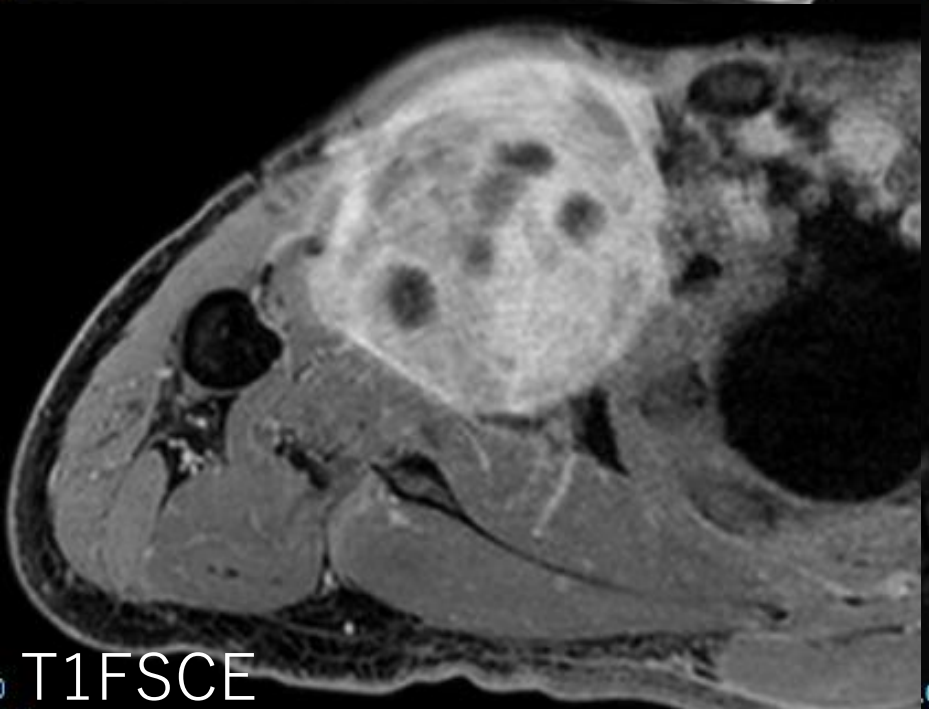


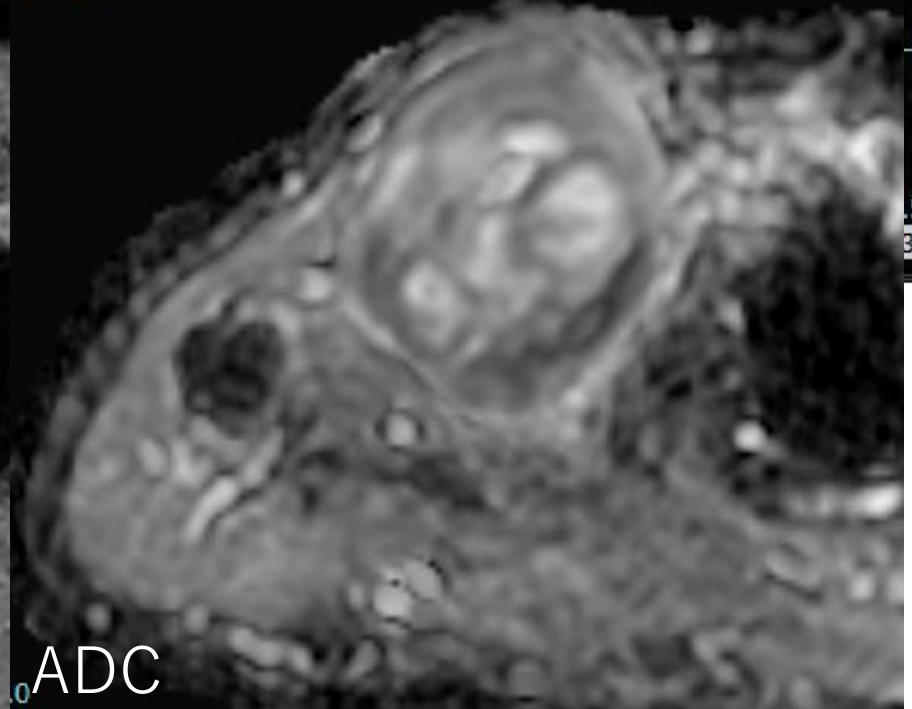
Mar, Year X+3
Before the initial resection











Oct, Year X+5
Recurrent tumor enlargement

症例2

帝京大学

Malignant Peripheral Nerve Sheath Tumor (MPNST) Presenting as Pancoast Syndrome in Neurofibromatosis Type 1

Patient: 36-year-old Female

Background: Diagnosed with Neurofibromatosis Type 1 (NF1). Positive family history (Mother and Maternal Grandfather).

Chief Complaint: Weakness and sensory disturbance in the left fingers (1-month duration).

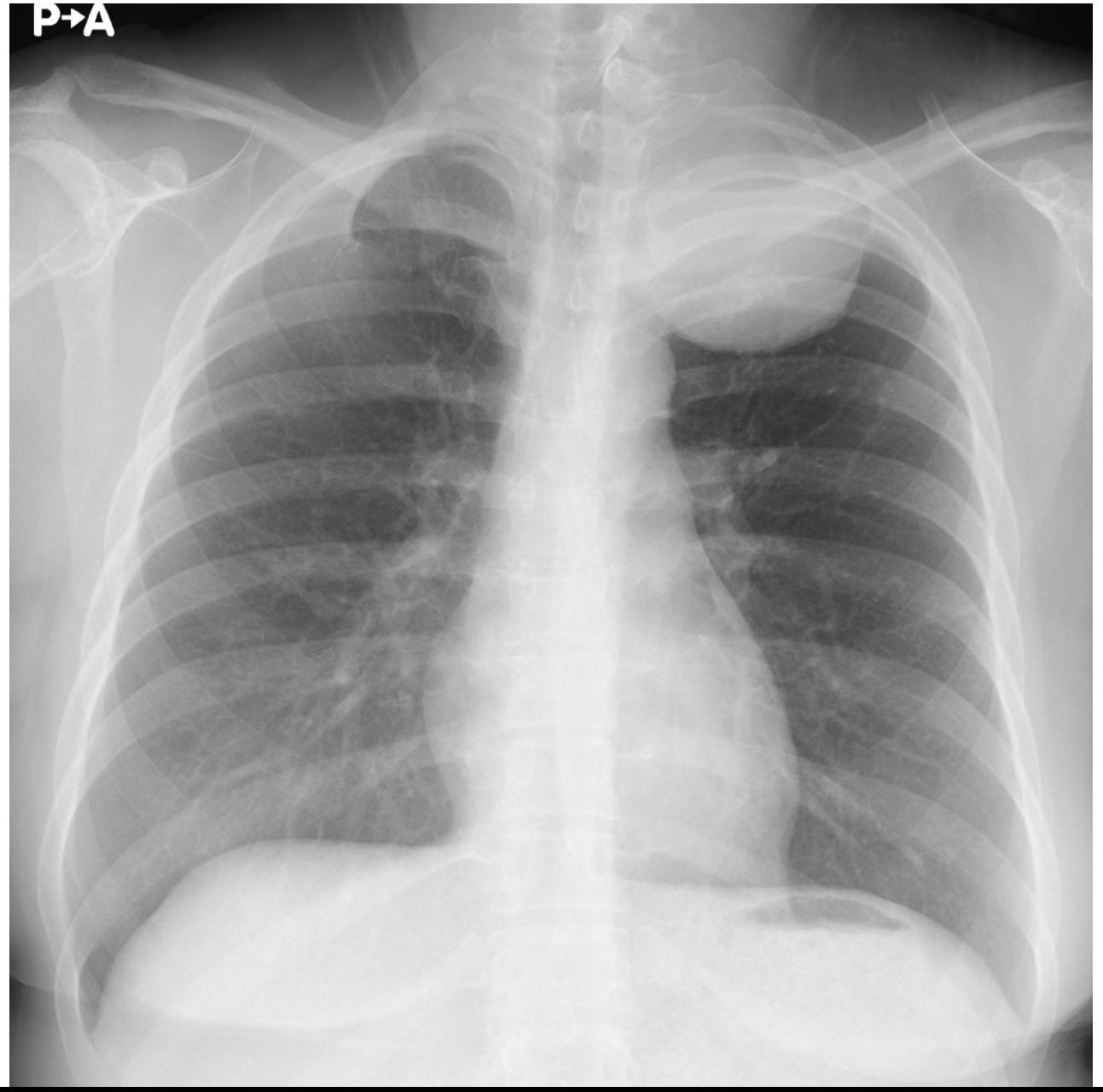
Physical Examination

Systemic Findings: Multiple neurofibromas and café-au-lait spots.

Horner's Syndrome: Presence of miosis, ptosis, and left-sided facial anhidrosis.

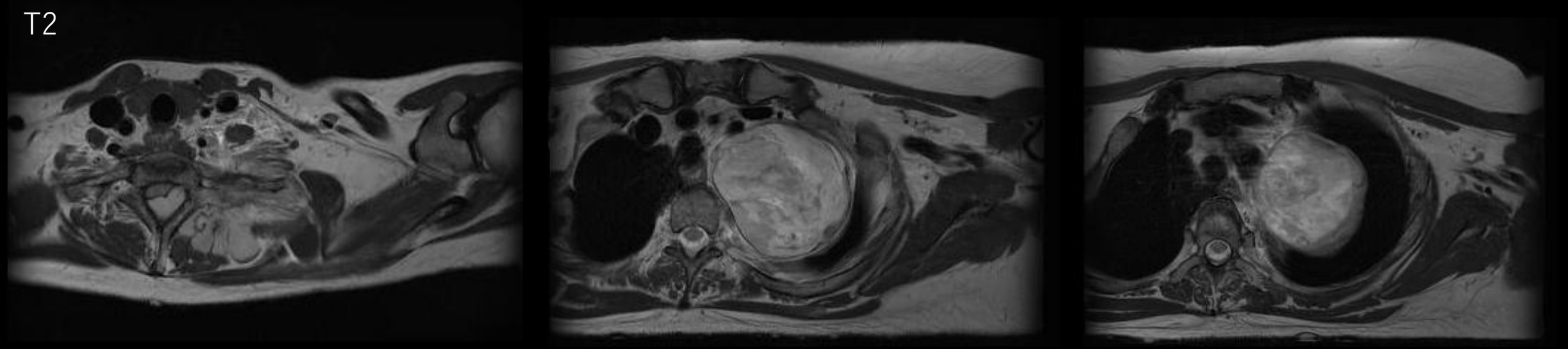
Neurological Deficits:

- Sensory loss in the left medial forearm and ring/little fingers (C8-T1 distribution).
- **Motor Weakness (MMT):** Wrist extension (4), Extensor digitorum (2), First dorsal interosseous (0-1), Abductor pollicis brevis (0-1).

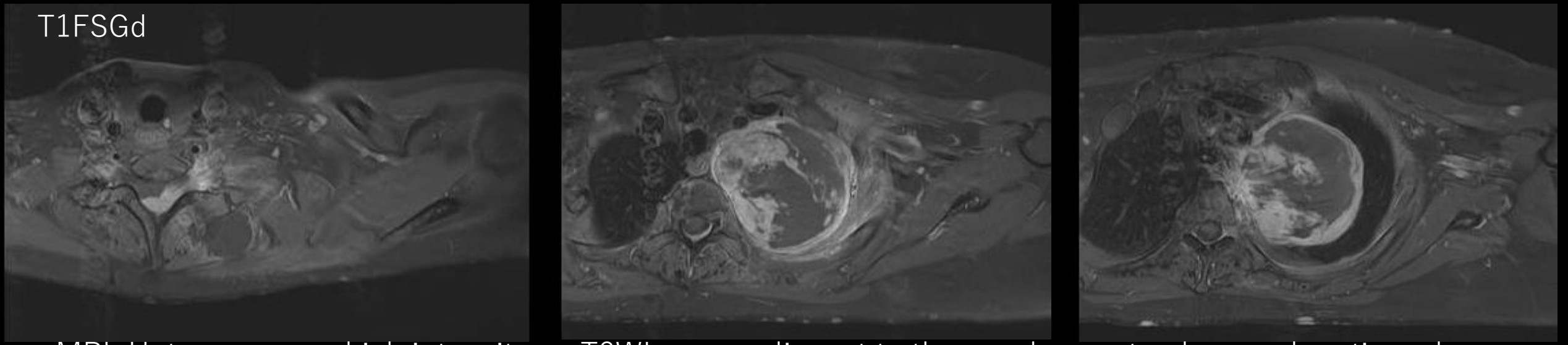


Chest X-ray: 8 cm well-defined mass in the left lung apex.

T2



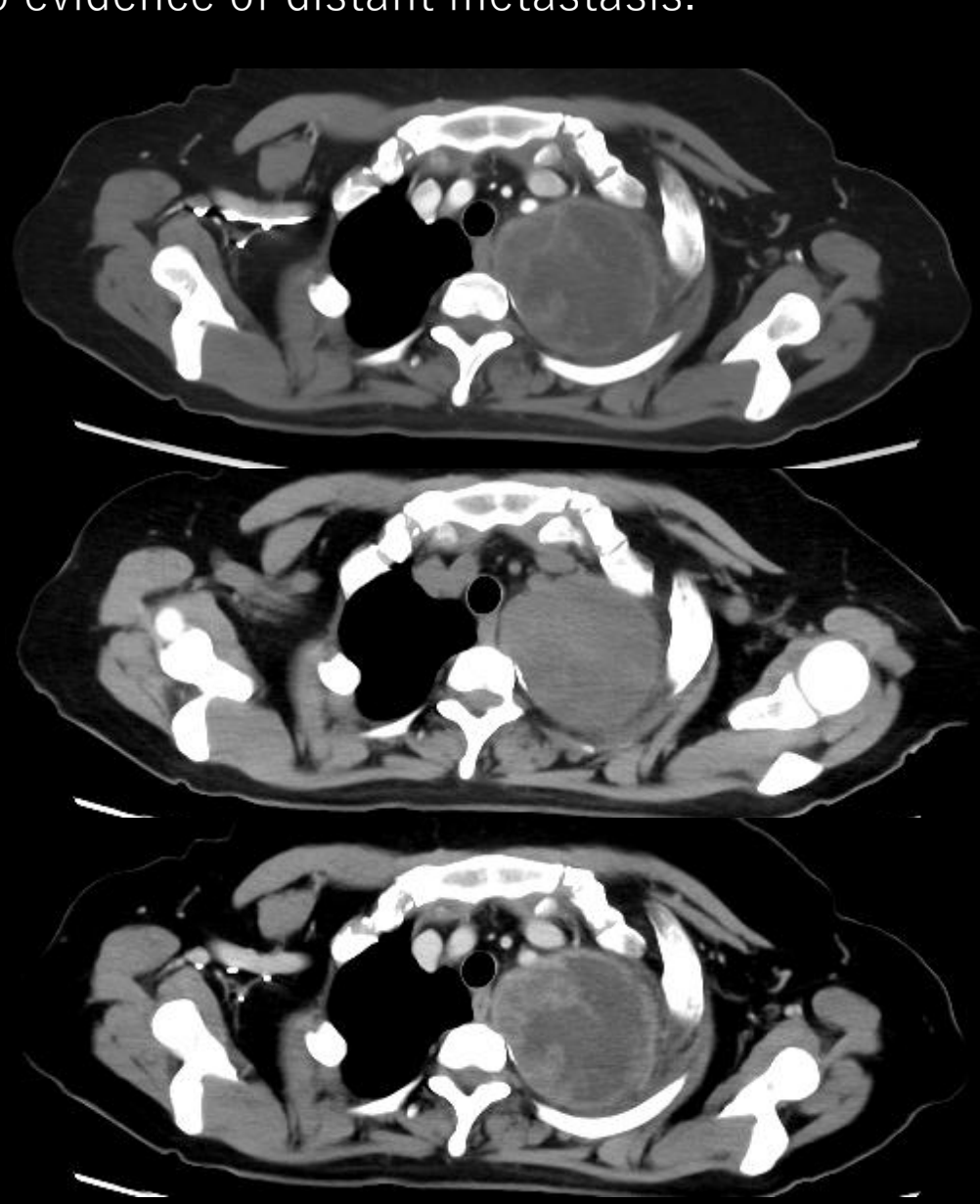
T1FSGd

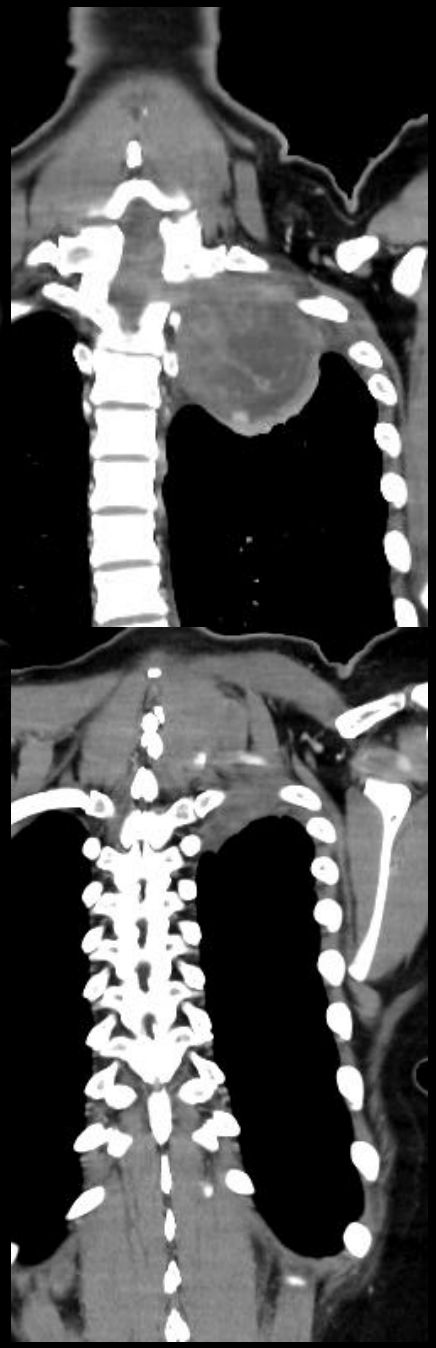


MRI: Heterogeneous high intensity on T2WI; mass adjacent to the esophagus, trachea, and aortic arch.

Dumbbell-shaped tumor: Continuous with the T1 nerve root, extending into the spinal canal via the neural foramen, causing spinal cord compression.

Staging: No evidence of distant metastasis.





Diagnosis: unresectable Pancoast-type MPNST arising from NF1

Treatment:

1. Surgical intervention (preparation for RT)

   Separation surgery: Partial resection of the tumor around the spinal cord to alleviate compression and create an anatomical margin for safe, high-dose radiotherapy.

2. Concurrent chemoradiotherapy (CCRT)

   Radiotherapy: 60 Gy / 30 Fr.

   Systemic chemotherapy: 4 cycles of AI Regimen (Adriamycin + Ifosfamide)

3. Maintenance therapy: transitioned to pazopanib following CCRT.

Clinical outcome:

Favorable course: this multimodal approach—integrating functional surgery, aggressive CCRT, and molecular-targeted therapy—achieved sustained tumor shrinkage.

Long-term stability: the patient remains in partial response (PR) 2.5 years post-treatment, demonstrating the efficacy of this combined strategy for unresectable MPNST.

症例3

静岡がんセンター

83-year-old female

Clinical History

Noticed masses in the mid-back and left shoulder 2 years before presentation

At presentation, referred to our hospital due to tumor growth

No personal or family history of Neurofibromatosis type 1 (NF1)

Physical Examination

Multiple cutaneous tumors, resembling NF1 phenotype

Mid-back mass : 14 × 10 cm

Left shoulder mass : 10 × 9.5 cm

PET-CT Findings

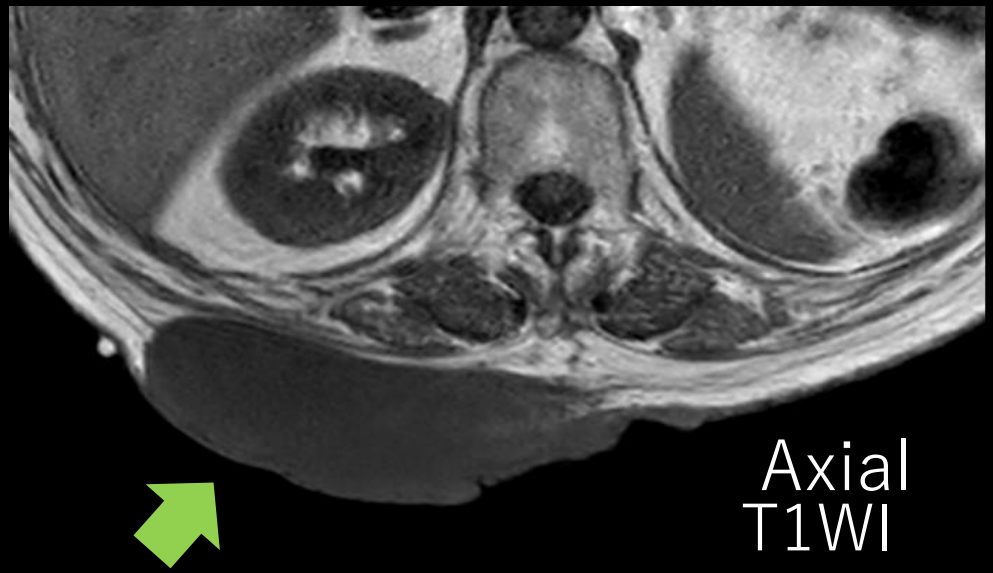
SUVmax : mid-back tumor 7.6, left shoulder tumor 17.3

Clinical Course

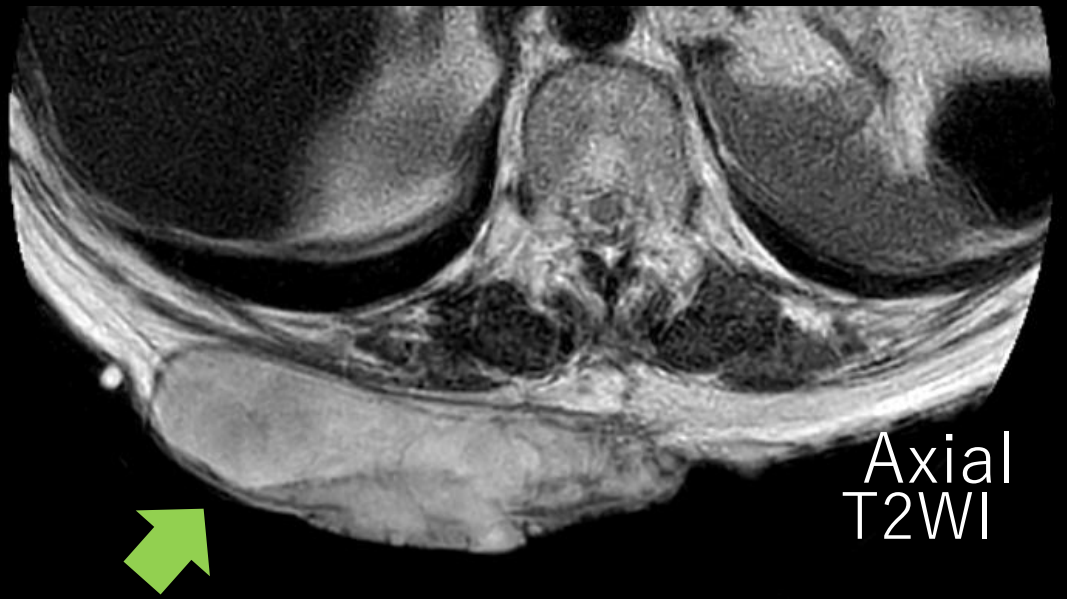
Core needle biopsy of both tumors at 2 weeks after first visit

Wide resection of both lesions at 3 weeks after biopsy

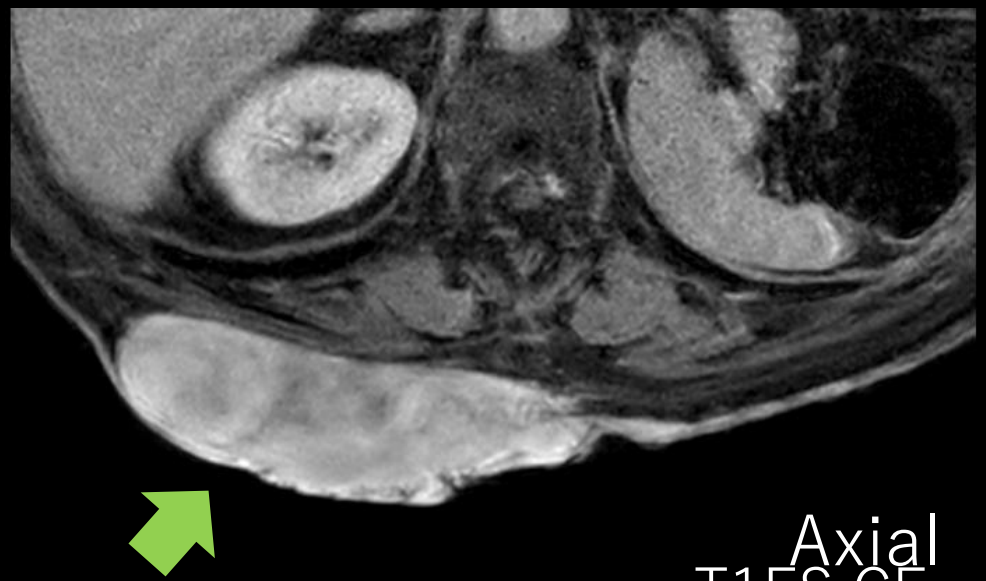
Mid-back tumor MRI



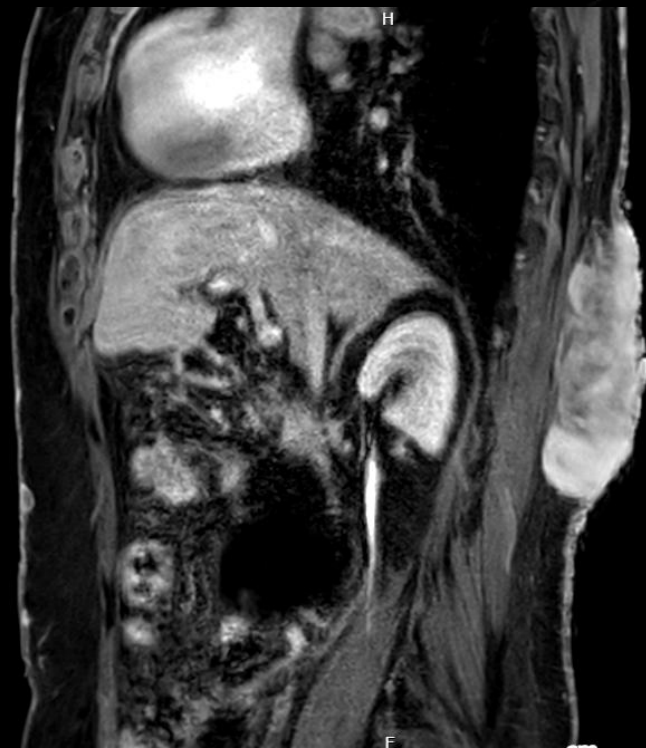
Axial
T1WI



Axial
T2WI

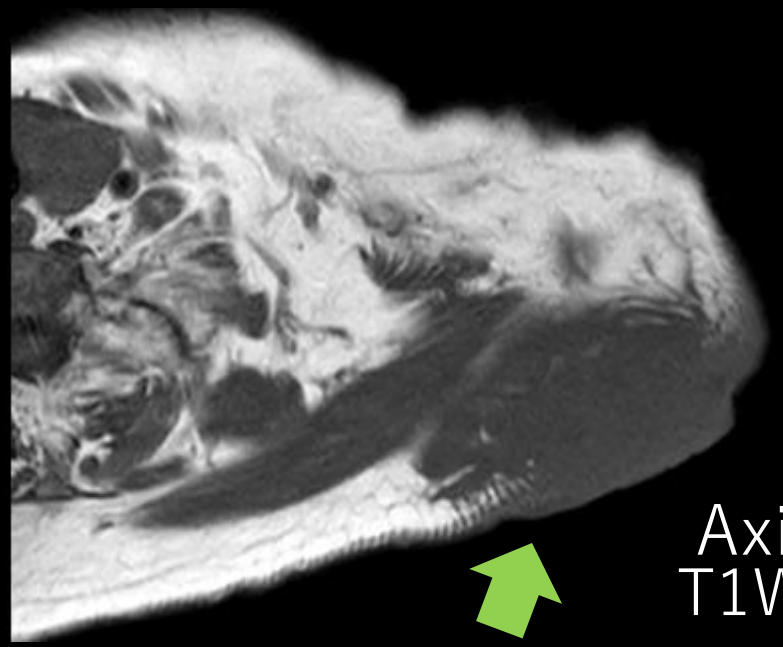


Axial
T1FS CE

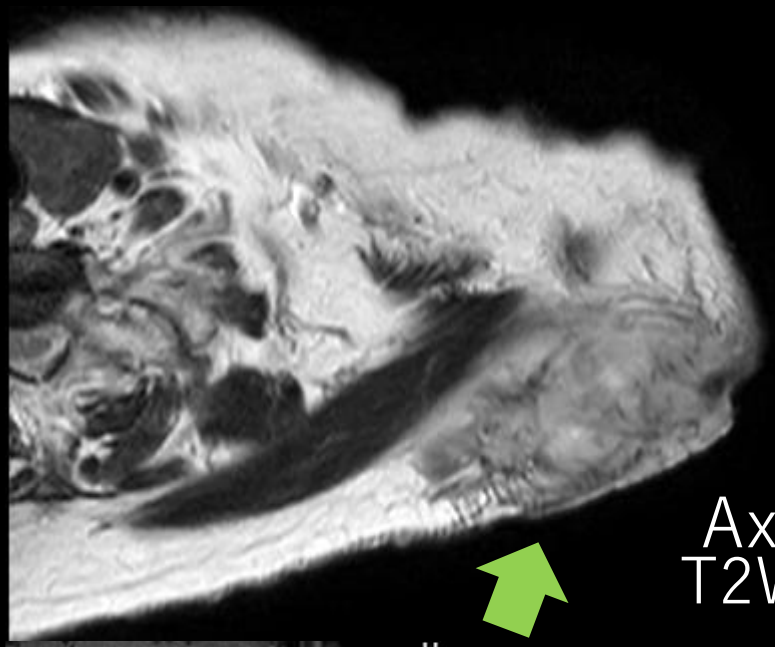


Sagittal
T1FS CE

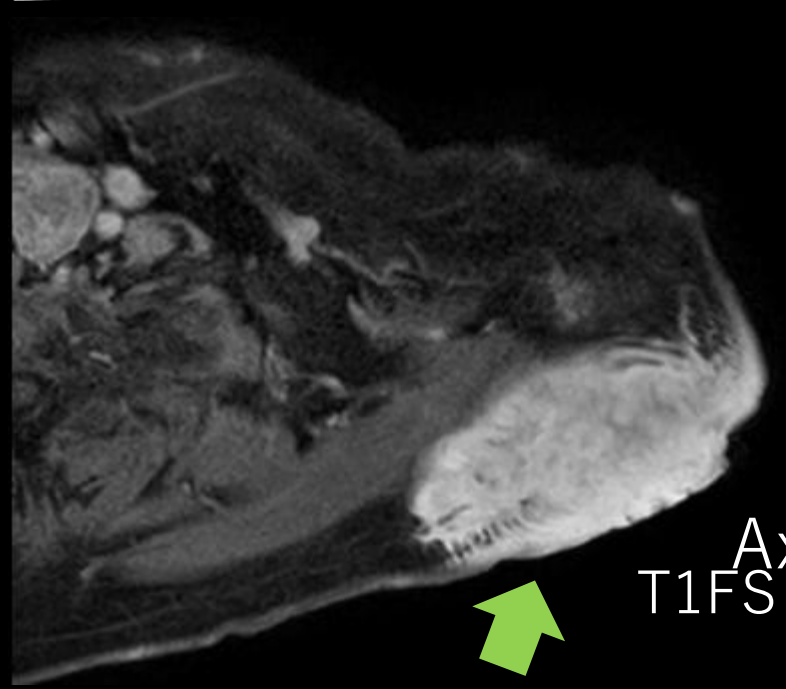
Left shoulder tumor MRI



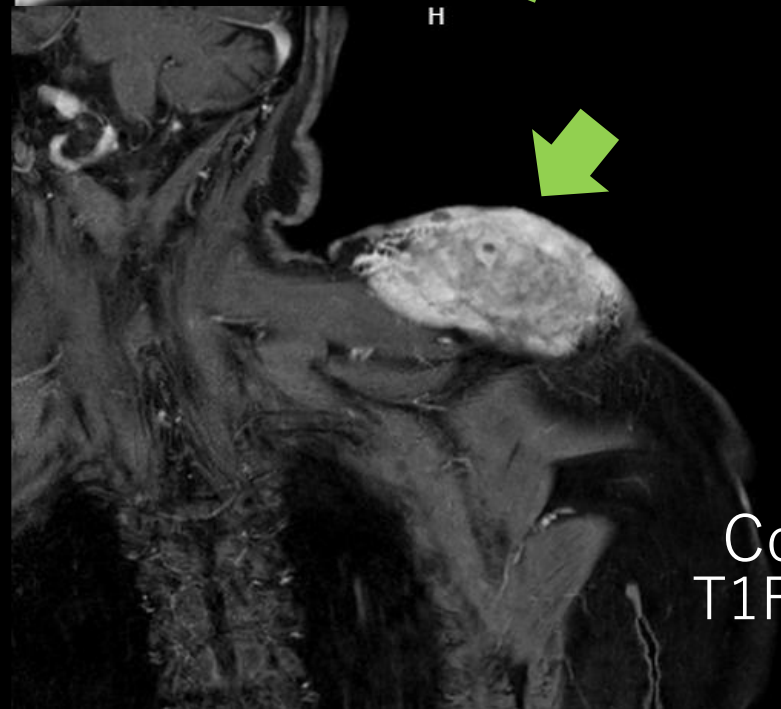
Axial
T1WI



Axial
T2WI



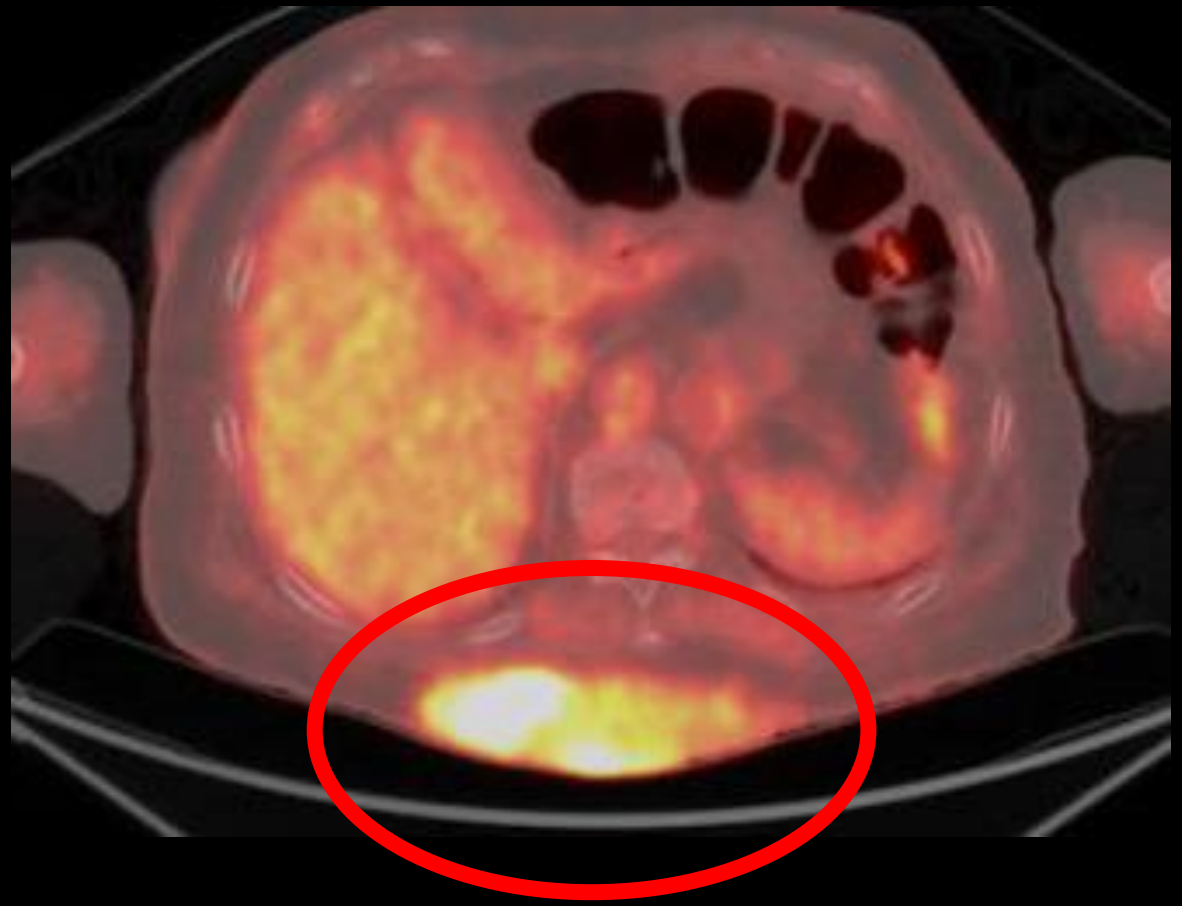
Axial
T1FS CE



Coronal
T1FS CE

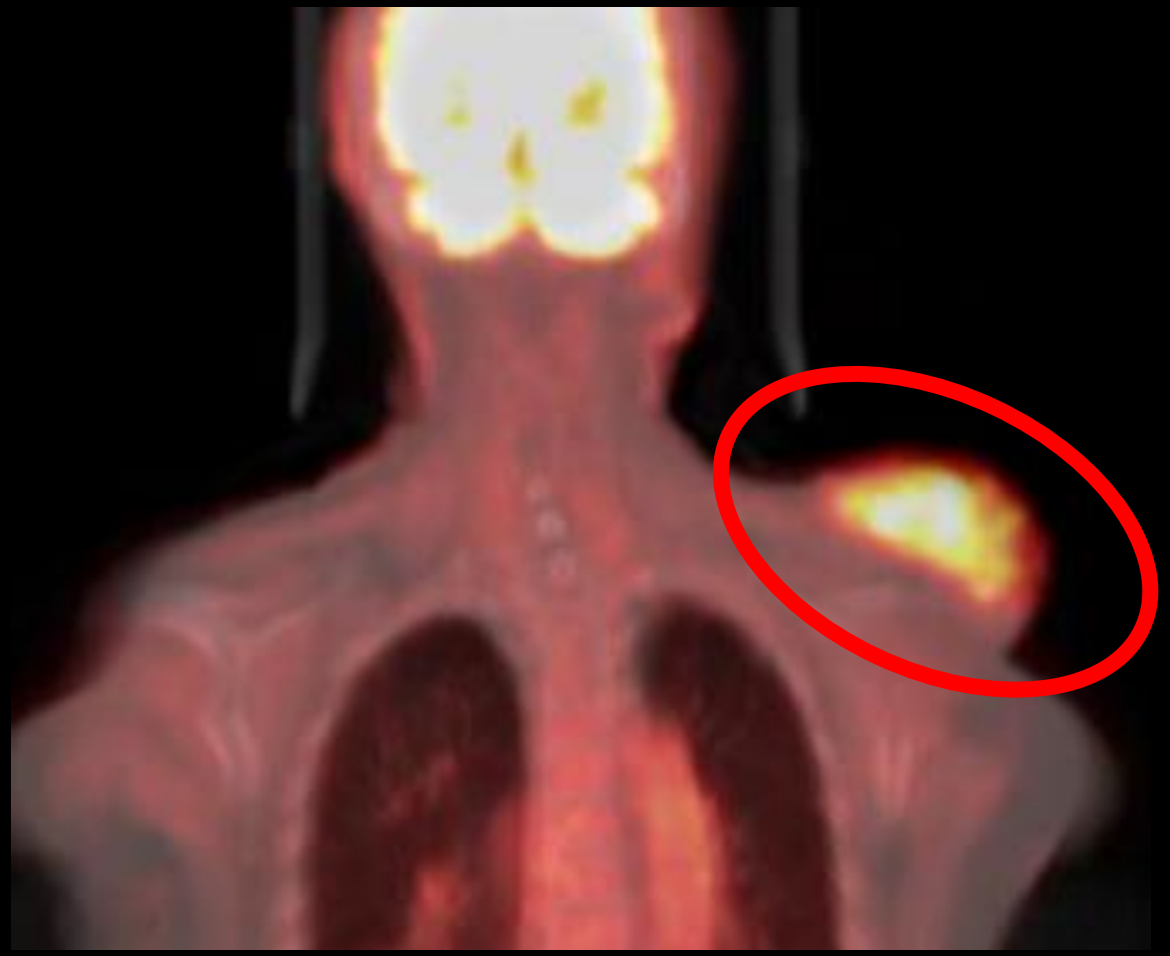
PET-CT

Mid-back tumor



SUV max 7.6

Left shoulder tumor

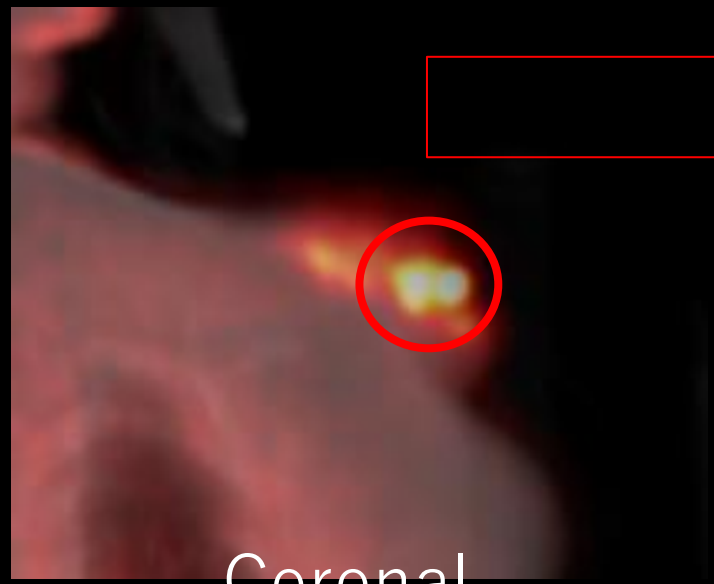


Left shoulder tumor

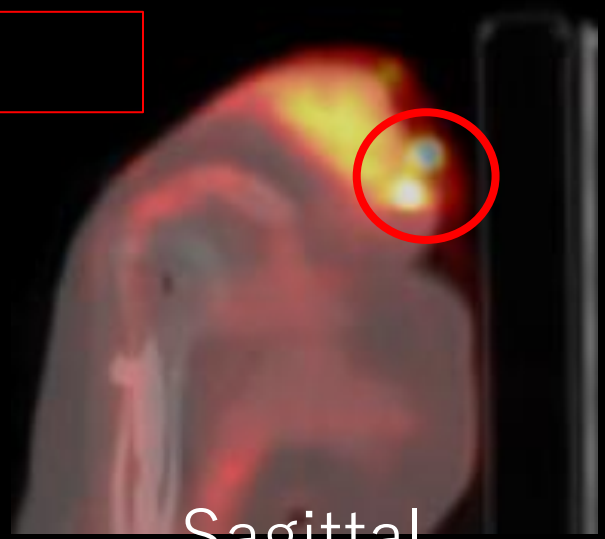
Only a small portion of the tumor's posterior lateral side shows very high uptake

PET-CT

Axial



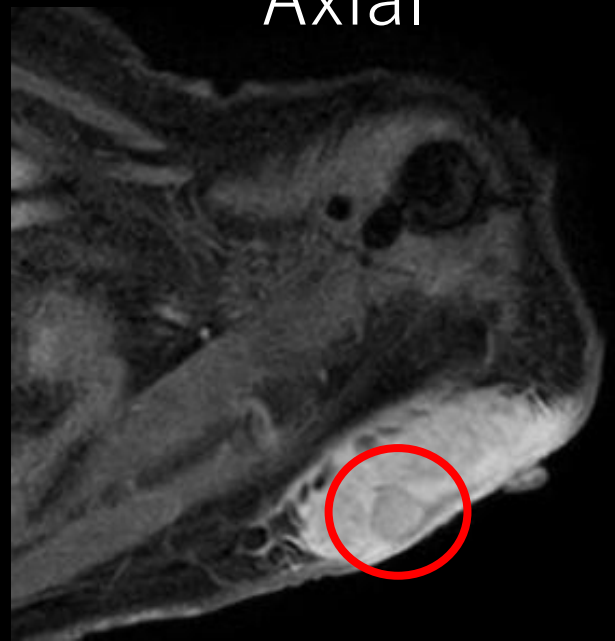
Coronal

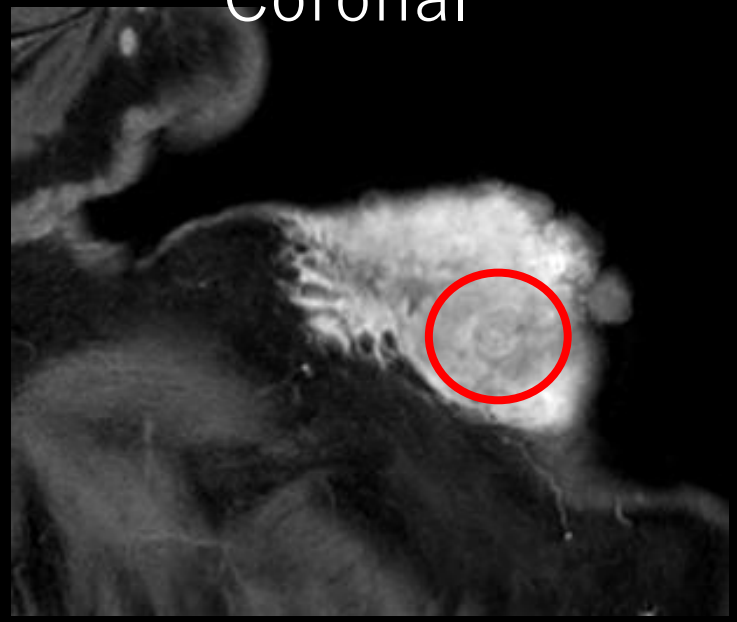


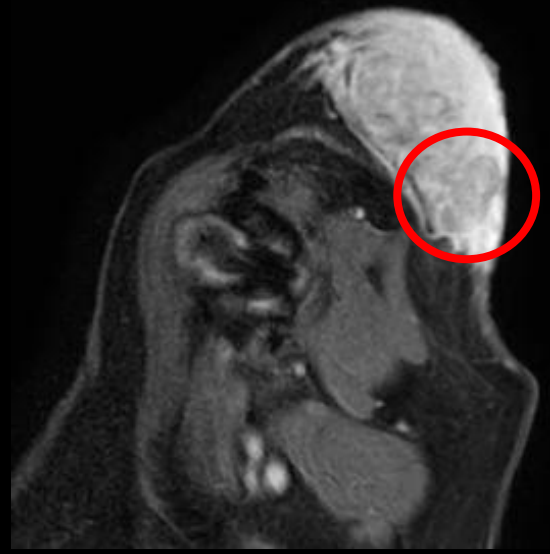
Sagittal

MRI

T1FS CE







症例4

がん研有明病院

A right chest wall tumor arising in a patient with neurofibromatosis type 1

Kyoko yamashita, Teruko Ueno, Norio Kurosawa,
Taisuke Tanizawa, Seiichi Matsumoto, Keisuke Ae

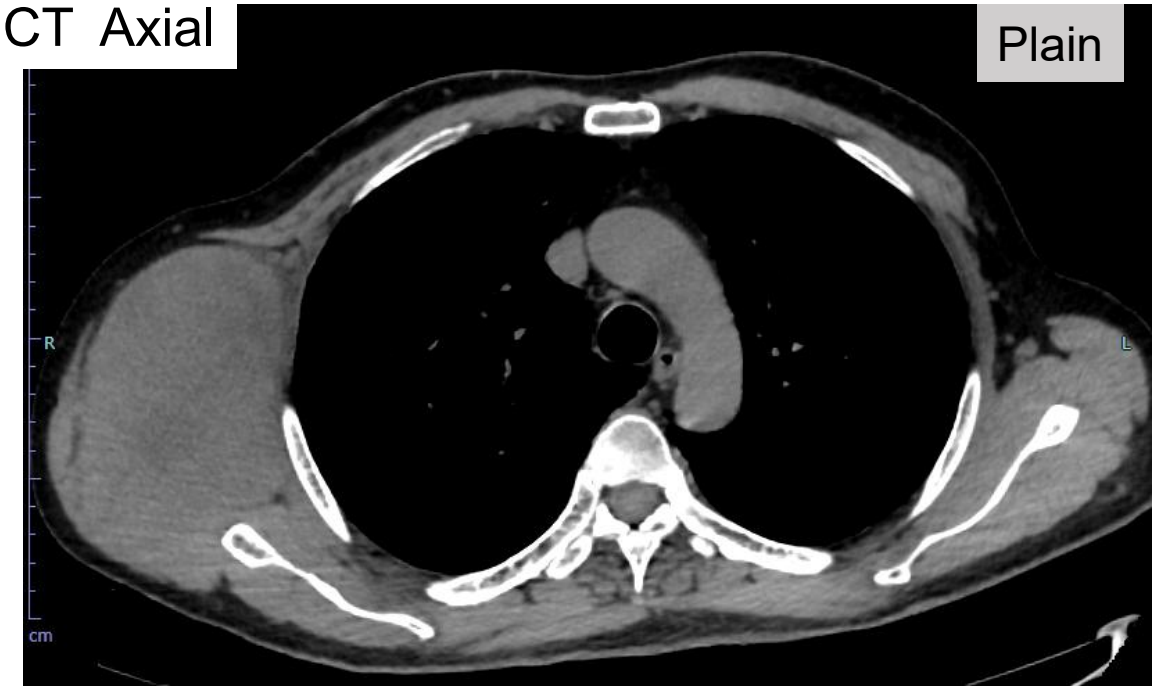
Case: 55-year-old male
Chief complaint: Right chest wall tumor

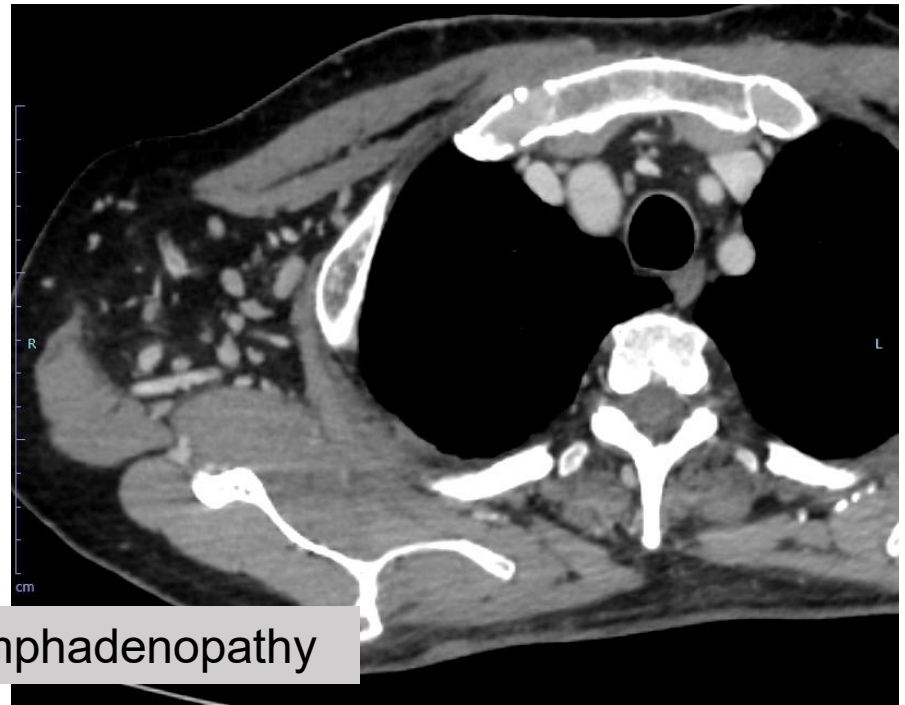
Background:
Multiple cutaneous neurofibromas and café-au-lait macules were present over the entire body, fulfilling the clinical diagnostic criteria for neurofibromatosis type 1 (NF1).

History of present illness:
Four months prior to presentation, the patient noticed a mass in the right chest wall (scapular region) while bathing. As the mass showed progressive enlargement, he visited a local clinic. MRI revealed an approximately 8 cm tumor located deep in the scapular region, and he was referred to our hospital.

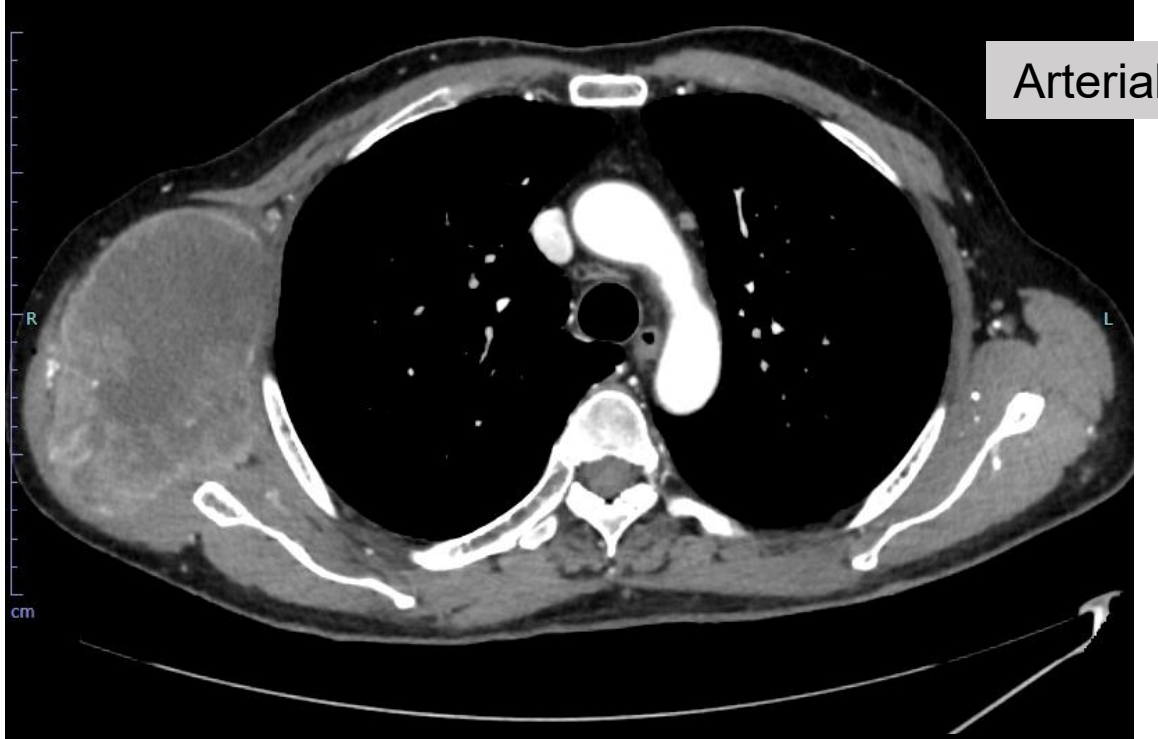
After needle biopsy, wide resection was performed.

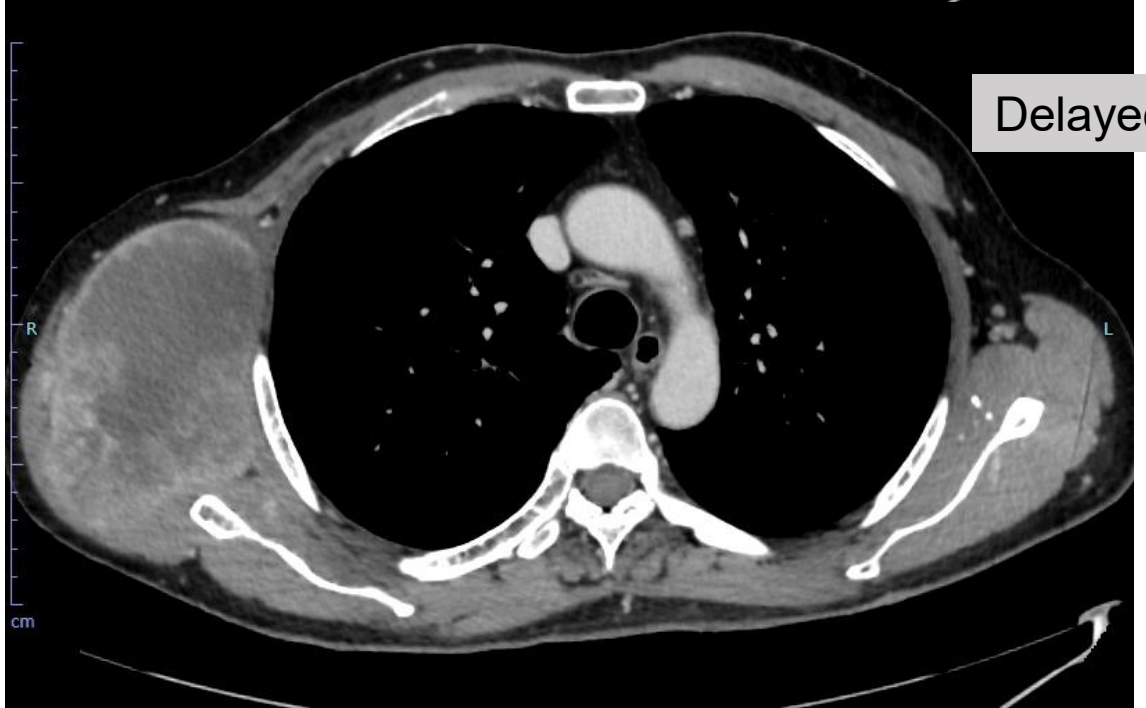
CT Axial





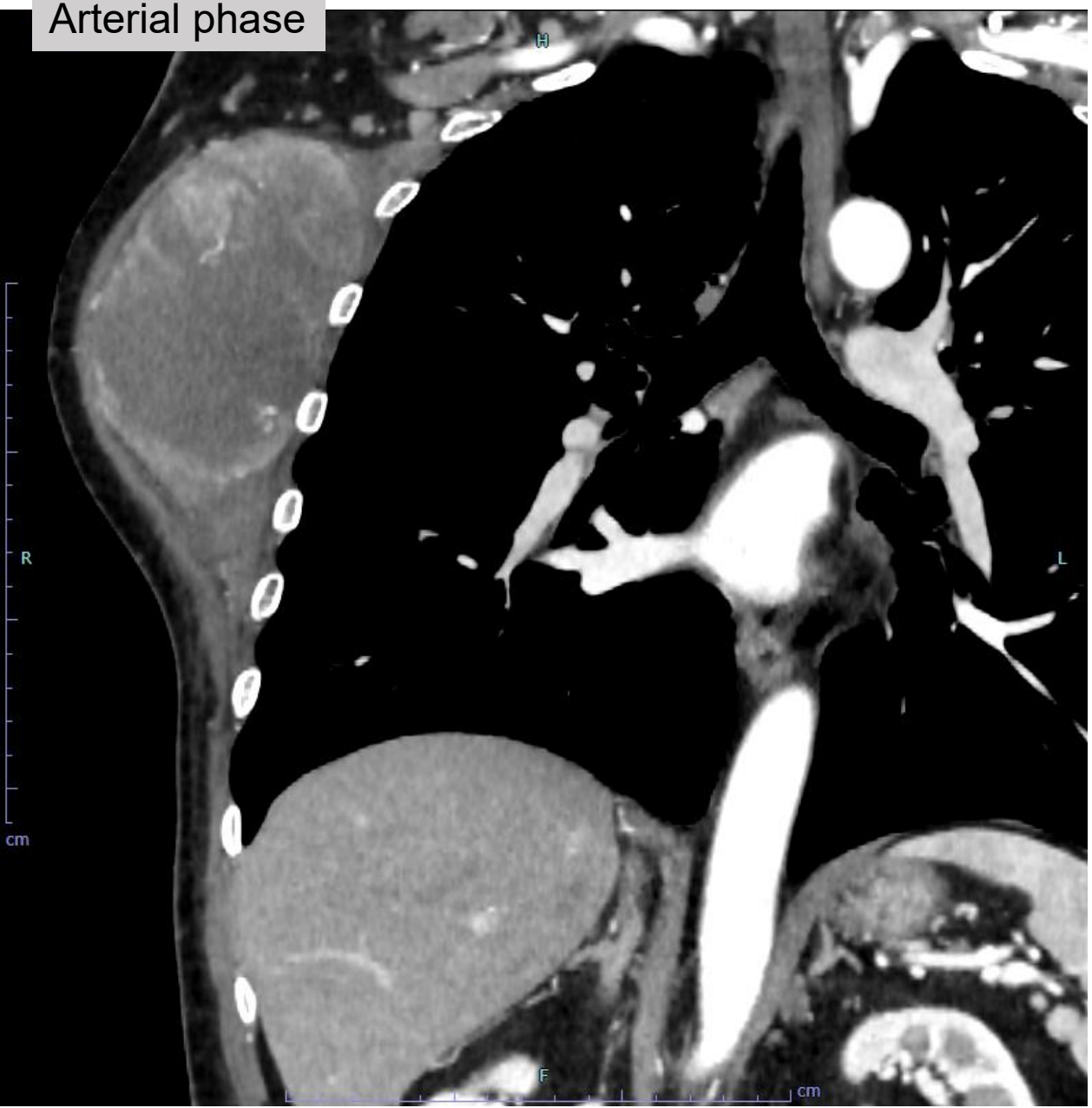
Axillary lymphadenopathy



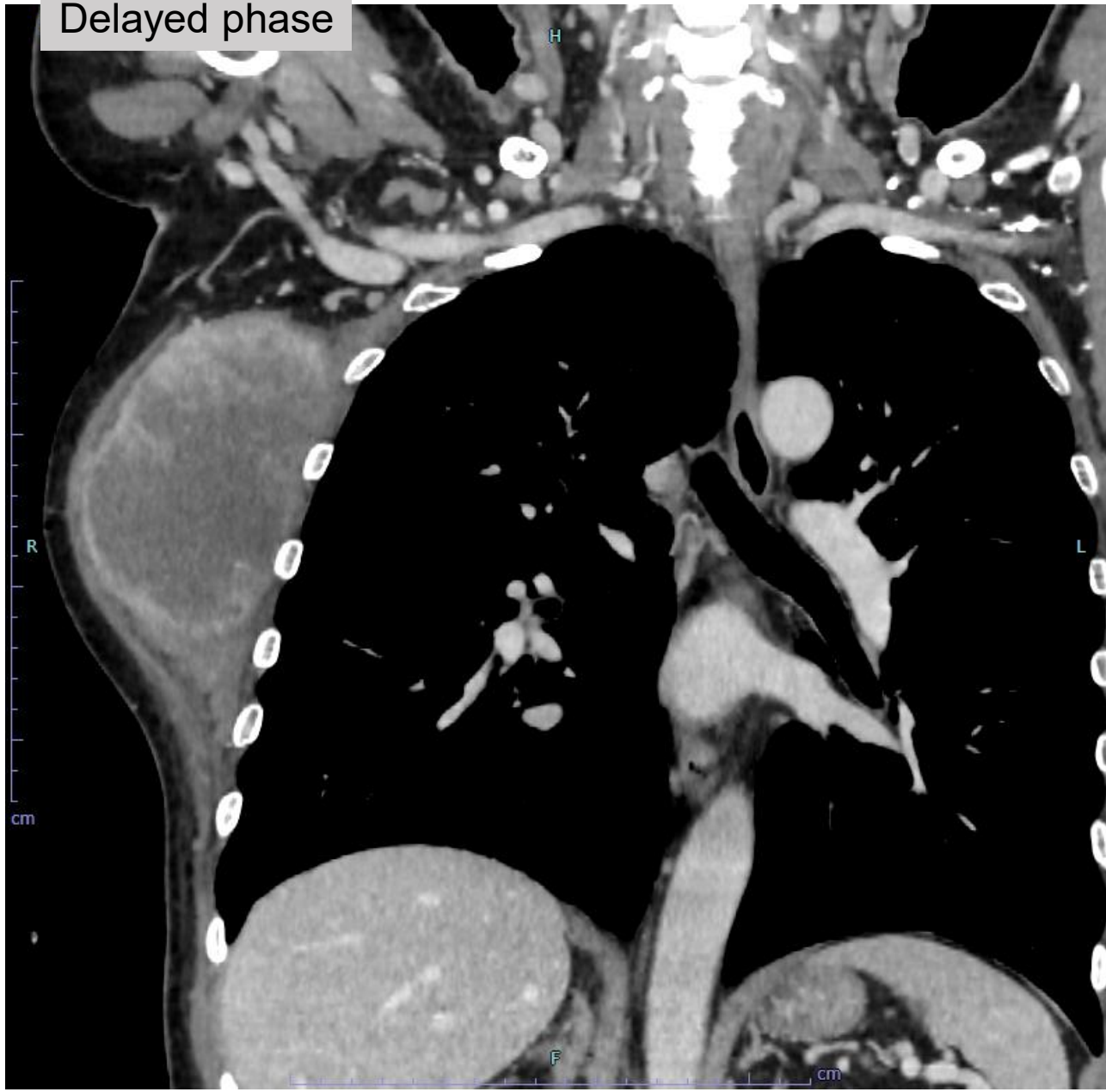


CT Coronal

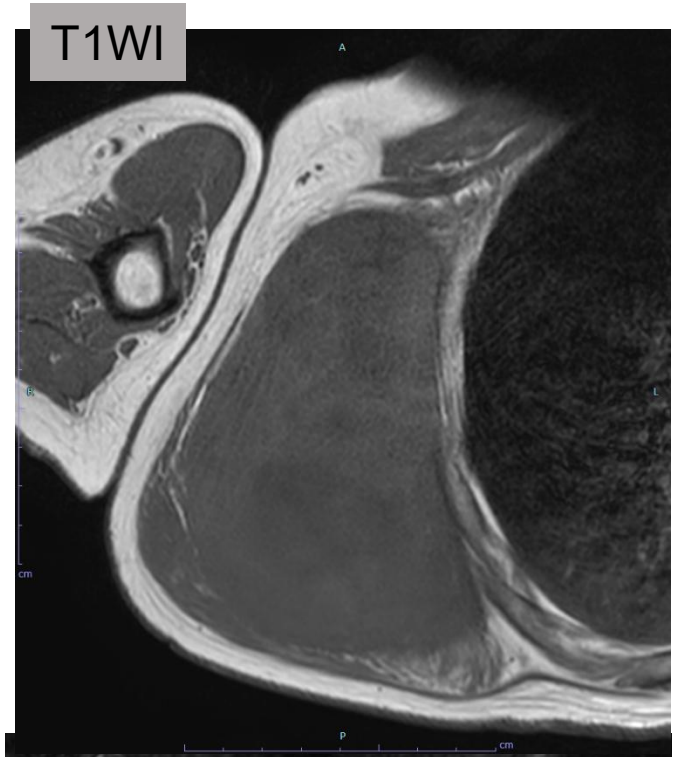
Arterial phase

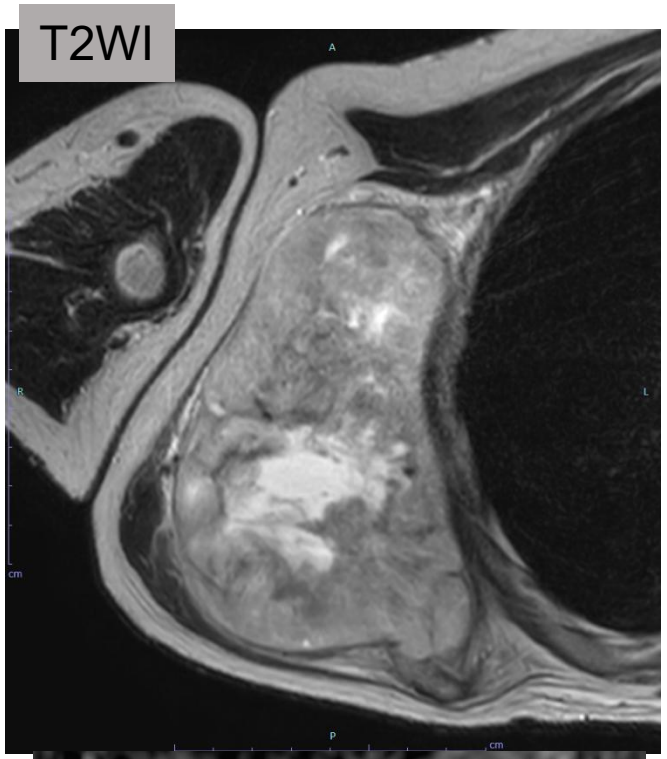


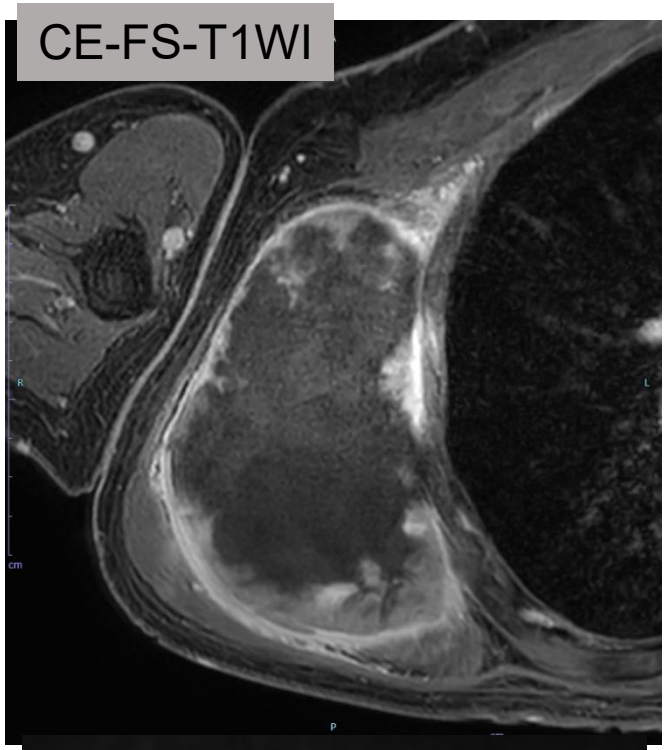
Delayed phase

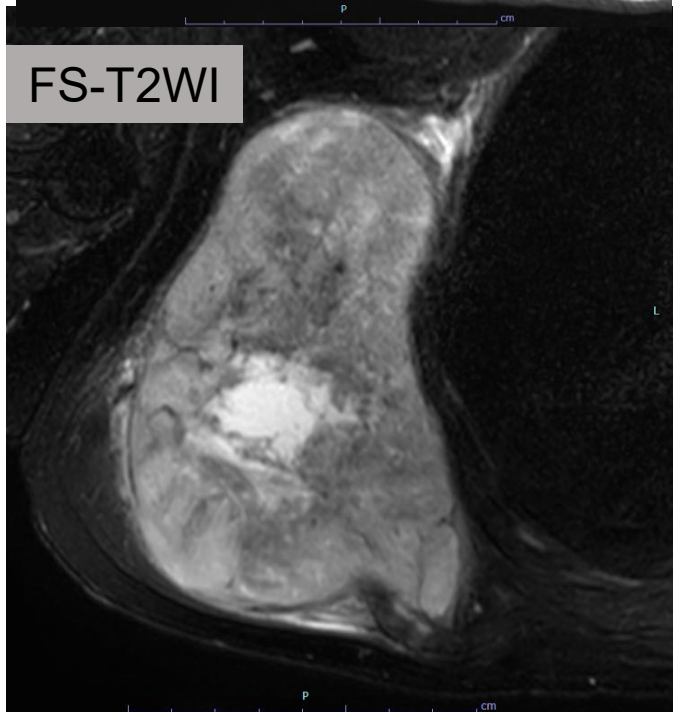


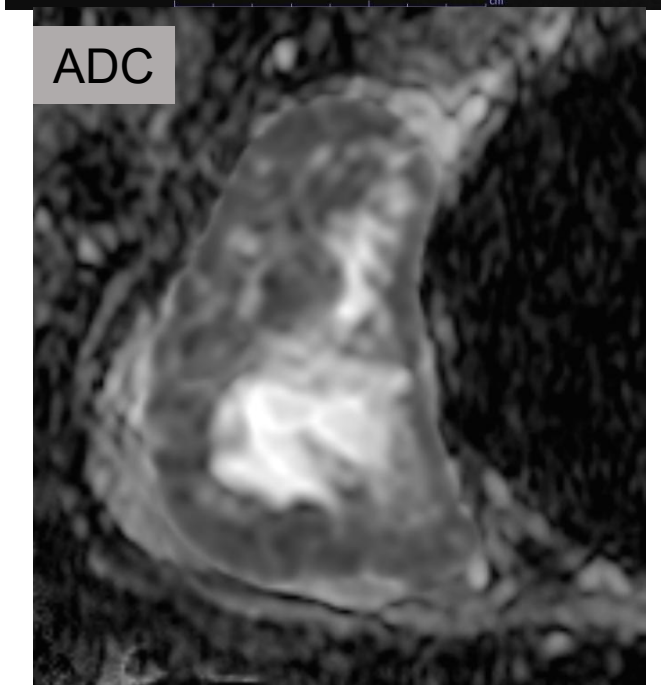
MRI Axial

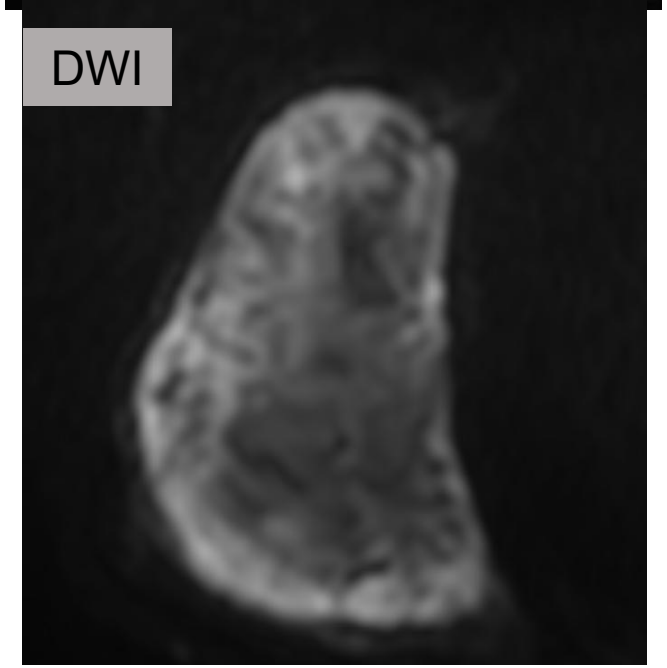












MRI Coronal