Penn Medicine

A case of pain and calcifications

Presented by: Mona Al Mukaddam, MD, MS
Associate Professor of Clinical Medicine and Orthopaedic Surgery
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Ian Cali Clinical Scholar at the Center for Research in FOP & Related Disorders

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Case Presentation

42YO M WITH CALCIFICATIONS

At age 11

- Presented with locked left knee
- Noted to have swelling and extensive calcium deposits
- Diagnosis of juvenile dermatomyositis was given
  - Steroids
  - Surgeries of his left knee and left arm to remove the calcium deposits
- Lost to follow up

In his 30's

- Progression of his calcifications, with pain in his right calf, right thigh, and right biceps causing mobility issues
- In June 2018 he underwent radical tumor resection on his right calf, pathology was consistent with tumoral calcinosis

Case Presentation - History

42YO M WITH CALCIFICATIONS

- In May 2022, he began to have right knee pain radiating to his hip which progressed over several months
- In February 2023, he underwent resection of two calcifying soft tissue masses of his R posterior thigh (15cm and 22 cm)
- Seen by Rheumatology and Dermatology who could not find evidence to support diagnosis of dermatomyositis
- No regrowth of excised lesions but continues to have flare ups of pain and significant mood symptoms. Most recently in February 2024 has developed left knee pain and swelling
- No prior history of trauma

Case Presentation

- **Medications:** Oxycodone-acetaminophen, Ibuprofen, Clonazepam, Vit D3
- **PMH:** Beta thalassemia trait, reported history of juvenile dermatomyositis, depression
- **PSH:** Left knee and left arm surgery (2011), Right calf mass resection (2018), R posterior thigh mass resection x2 (2023)
- **SH:** No tobacco or alcohol use, + marijuana. Currently not working
- **Family History:**
  - Daughter, age 16, healthy Son age 8 healthy
  - Mother: chronic pain of unknown etiology,
  - Father: CAD, prostate cancer with bone mets
  - Brother: healthy

Case Presentation

PHYSICAL EXAM:

- VS. BP 115/74 HR 72. Ht 5’5’’ Wt 157lb. BMI 26.16

- **Gen:** No acute distress, walking with cane
- **Skin:** Right LE surgical scar, L knee surgical scar
- **MSK:** R bicep palpable soft mass ~4cm, R posterior fossa (knee) with soft lump, hard lesions in R posterior thigh
- Normal toes and fingers
- No joint swelling or erythema

Biochemical Evaluation

Ca	Alk Phos	Vit D 25OH	PTH	Mg Phos	Cr eGFR	Other
9.3	80	14 (L)	6.1	2.2 3.5	0.91	TSH 0.94 Uric acid 6.5 SPEP normal Hgb 13.3, MCV 64

Myositis Extended Panel – all negative

SSA 52 Ab
SSA 60 Ab
Anti RNP
Jo-1 ab
PL-12 (alanyl-tRNA synthetase) Ab
PL-7 (threonyl-tRNA synthetase) Ab
EJ (glycyl-tRNA synthetase) Ab
OJ (isoleucyl-tRNA synthetase) Ab
SRP (Signal Recognition Particle) Ab
Ku Ab
PM/Scl-100 Ab
Fibrillarin (U3 RNP) Ab
Mi-2 (nuclear helicase protein) Ab
P155/140 (TIF1-gamma) Ab
SAE1 (SUMO activating enzyme) Ab
MDA5 (CADM-140) Ab

Genetic Testing

– WHOLE GENOME SEQUENCING NEGATIVE

Test(s) Requested

Diagnostic Testing / GenomeSeqDx / Clinical Genome Sequence Analysis

Clinical Indication

Individual with heterotopic ossification and hypertension.

Result: Negative

- Causative variant(s) in disease genes associated with reported phenotype: **None Identified**
- Variant(s) in genes possibly associated with reported phenotype: **None Identified**
- ACMG Secondary Findings: **None Identified**
- mtDNA Test Results: **Mitochondrial DNA sequencing and deletion results are reported separately.**

Pathology Results

2018

Right calf mass:

Fragments of soft tissue with a calcifying shell and central accumulation of partly calcified amorphic eosinophilic material, consistent with tumoral calcinosis

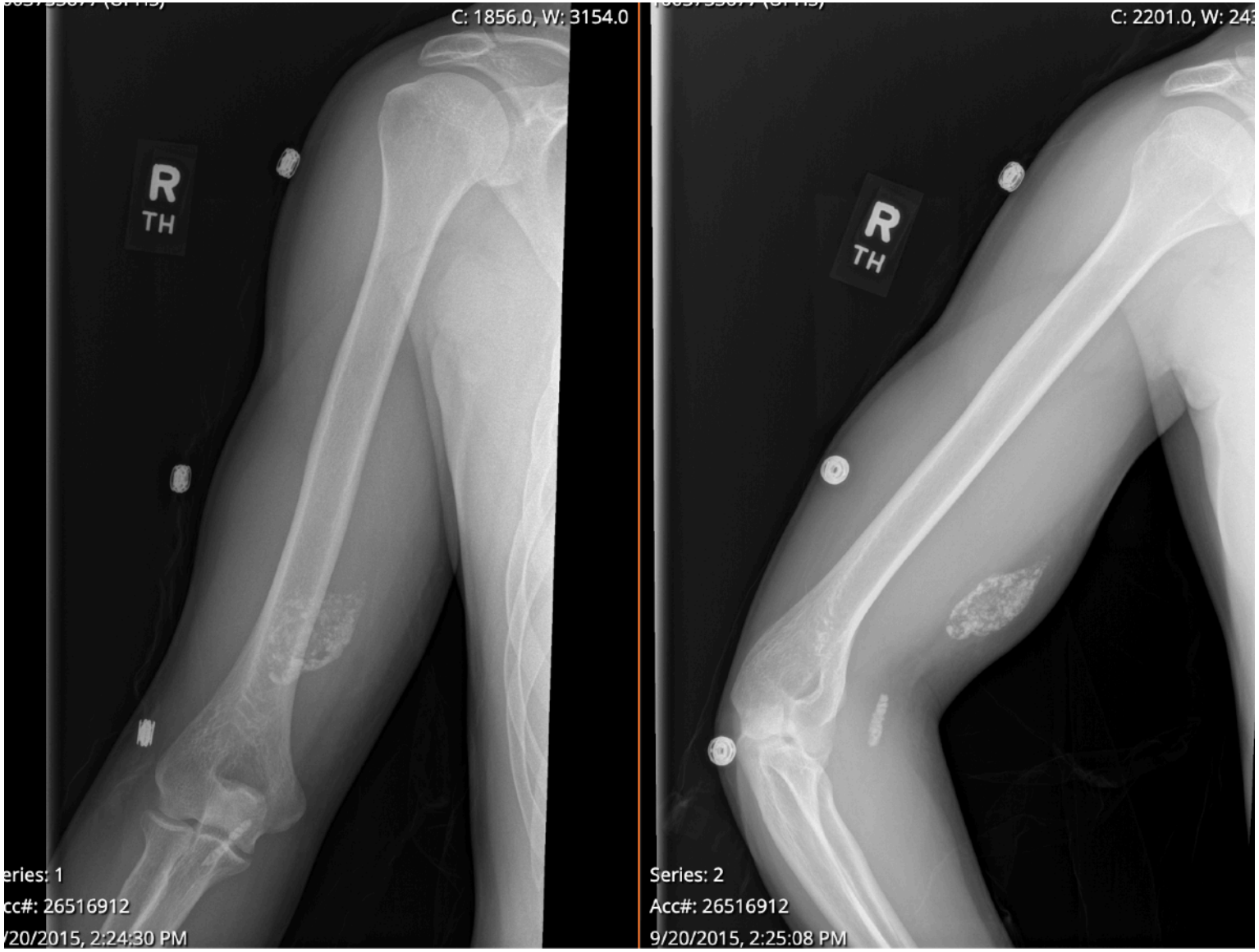
2023

Right posterior thigh mass and right posterior knee mass:

Cystic spaces separated by thick fibrous septae lined by histiocytes and multinucleated giant cells and filled by amorphous granular material. Focal osseous metaplasia seen

Imaging

– R humerus



Imaging

– R tibia/fibula

Imaging

– R Femur

Imaging

– R Femur

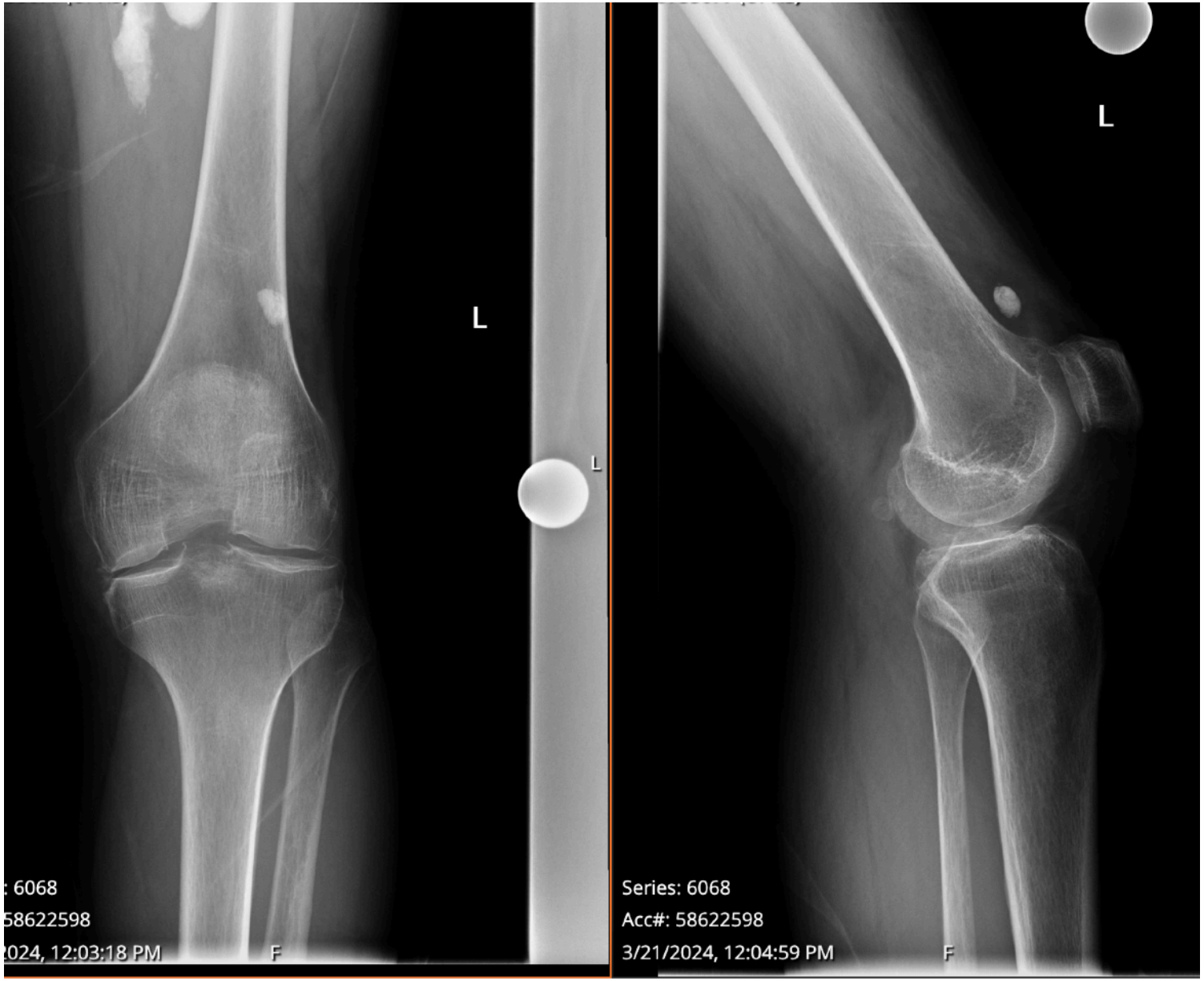
Imaging MRI 2022

Interval progression of soft tissue masses/calcium-deposition process involving the right thigh posterior thigh soft tissues and hamstring musculature compared to prior study dated 6/13/2022. New STIR-hyperintense signal in the two known distal biceps femoris greater than semitendinous lesions which show stable to slight increased size. Multiple new lesions with likely thin rim calcification and internal fluid-calcium/hemorrhage levels. There is peripheral enhancement and surrounding soft tissue/enhancement. The overall process spans approximately 30 cm x 8.7 cm transverse x 3.1 cm AP from the level of the proximal-to-mid femoral diaphysis to the femoral metadiaphysis.

Differential could include myositis ossificans progressiva, tumoral calcinosis, or other etiology. Correlate with patient history.

Imaging

– L Knee



Imaging - DXA

LUMBAR SPINE	FEM NECK	FEM TOTAL
BMD 0.977. Z -2.0 TBS 1.236	Left BMD 0.723. Z -2.4 Right BMD 0.782 Z -1.9	Left BMD 0.732. Z -2.4 Right BMD 0.737 Z -2.3

Clinical Questions

What is the diagnosis?

Would there be any role for molecular testing of the calcified lesions?

Is additional surgery recommended?

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Thank you

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