



6th International ECD Medical Symposium

NOVEMBER 2018

ORLANDO, FLORIDA

Case Description

- ▶ 40 y.o female seen in July 2018 by us for management of ECD
- ▶ 2008 bone cyst in RIGHT tibia treated with a graft, followed by knee pain and swelling after that
- ▶ Diagnosed with DI in 2008: on desmopressin
- ▶ 2013: swelling left ankle with bone cyst: treated with excision of tendon sheath mass. Pathology shows inflammation with lymphocytes and histiocytes-some multinucleated
- ▶ 2014: Right knee synovial biopsy: villous hyperplasia with inflammation
- ▶ 2016 developed LEFT knee pain and new cyst in the distal femur, treated surgically
- ▶ 2016 Pathology c/w chronic synovial hypertrophy with giant cells
- ▶ 2016 pathology consultation: c/w Tenosynovial giant cell lesion with prominent xanthomatous component, favoring TENDINOUS XANTHOMA on specimen from 2013
- ▶ 2017: resection of posterior knee mass and synovial lesion of left knee: c/w chronic synovitis with moderate synovial proliferative features

Case Description

- ▶ PET/CT 2016:
 - ▶ b/l knee effusions with synovial thickening and MILD hypermetabolic activity
 - ▶ Complex cystic bone lesions in b/l tibiotalar joint, b/l popliteal fossa, with MILD hypermetabolic activity
 - ▶ Splenomegaly
- ▶ Treated by rheumatology for possible inflammatory arthritis of lower extremities with multiple drugs, without success and gradual worsening of pain
 - ▶ Hydroxychloroquine
 - ▶ Corticosteroids
 - ▶ Oral methotrexate
 - ▶ Adalimumab (Humira)
 - ▶ Etanercept
- ▶ Significant PMH/PSH/FH: xanthelasmas since her 20s

At the time of the consultation:

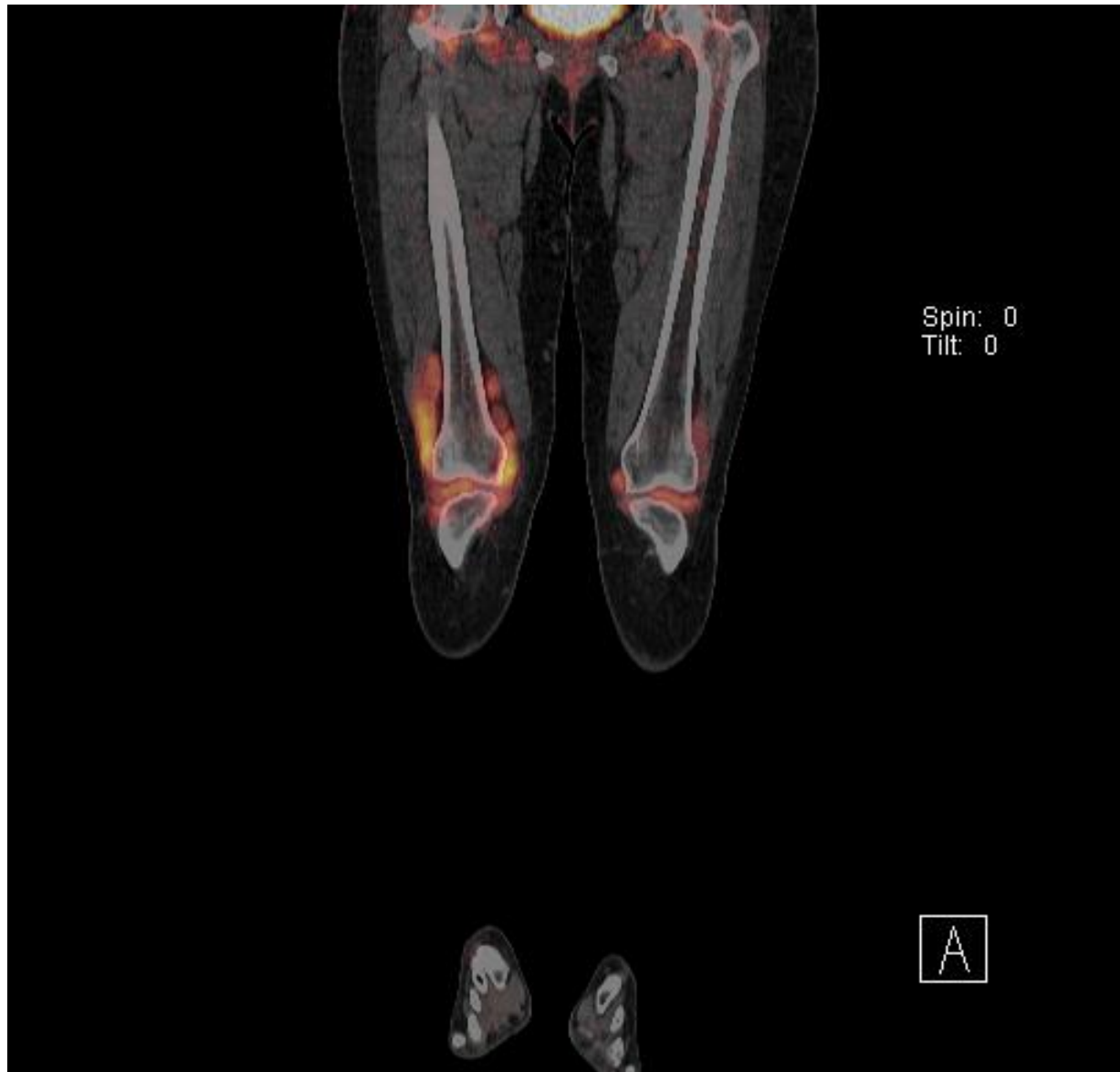
- ▶ Additional pre-consultation information: skin biopsy (Xanthelasmas) c/w foamy histiocytic infiltrate with non-LCH features (CD1a and S100 negative, FXIII +/-)
- ▶ The patient self referred after consultation with rheumatology at Mayo Clinic Jacksonville where treatment with Anakinra was recommended
- ▶ Main complaints: moderate to severe b/l knee pain, fatigue, diaphoresis, possible nocturnal fevers
- ▶ Significant PE findings: bilateral xanthelasmas 4-5 cm, b/l knee effusions and chronic synovitis changes, limited ROM of knees
- ▶ BRAF V600E studies: tissue and cfDNA: negative
- ▶ Medications: tramadol and desmopressin

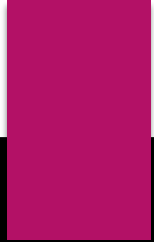
Imaging done after consultation (2018)

- ▶ Significant findings
 - ▶ MRI of brain: Depression of the pituitary tissue, with asymmetry, lack of pars posterior bright spot
 - ▶ PET/CT:

Clinical course after consultation

- ▶ 6 week course of anakinra: no change in inflammation/pain
- ▶ Started Trametinib (Mekinist) 2mg/day OCT 5 2018
 - ▶ 1 week follow up: less fatigue, mild nausea
 - ▶ 4 weeks later: less fatigue, less knee pain, resolution of diaphoresis/nocturnal fever, increased ROM





Spin: 0
Tilt: -90

