



# 4/25/26 Simplicity in Complexity w/ @CPSolvers

*"One life, so many dreams"* Case Presenter: Jeffrey Case Discussants: Kirtan (@KirtanPatolia)  
<https://clinicalproblemsolving.com/present-a-case/>



Scribing (Sarah B)

## *"The Shapeshifter"*

**CC:** 32 yo M with rheumatoid arthritis in setting of a febrile illness.

### HPI:

Several members of the household tested positive for Covid with fever cough and sore throat. He had these symptoms for 1 week followed by arthritis and body aches. In the past year has had joint pain and swelling in PIPs and morning stiffness. Due to symptoms, presented to PCP who ordered a rheum panel. ESR 52 but ENA/other tests were negative. He was referred to rheumatology at this time. On rheum exam PIP and of fourth and fifth fingers showed synovitis. No malar rash or other skin findings. HLA-B27 negative. Uric acid elevated. XR of joints were negative. Was started on methotrexate and allopurinol. Dual energy CT to identify gout was ordered and negative for gout, so tx was stopped. Seronegative RA diagnosis was made. MTX and folic acid were current meds at initial presentation.

### ROS:

Rheumatologic ROS all negative except easy bruising despite normal coags/plts.

### PMHx

Recurrent Tonsillitis as a kid  
Legg-Calvé-Perthes disease  
Chronic or recurrent sinusitis with eosinophilia  
Septal deviation s/p ENT surgical correction  
Recurrent respiratory infections (last 6mo)  
ITP in childhood, resolved with steroids

### Hospitalization #1

**Vitals:** T: 100.4 F HR: 100 BP: nl RR: nl Sat: nl

**Exam:** HEENT: nl CV: nl Pulm: nl Abd: nl Neuro: nl Extremities: L knee effusion, warm and tender to palpation.

### Notable Labs & Imaging:

#### Hematology:

WBC: 4.7 (N66%) Hgb: 11.5 Plt: 530K MCV: 112

Folic Acid nl, B12: 6 (L)

#### Chemistry:

BMP normal AST: 52 ALT: 26 Alk-P: nl

Arthrocentesis: 25K WBC, CPPD crystals

Infectious testing negative (e.g. Lyme, GC/CT (triple site))

RF, CCP etc. negative, Uric acid elevated, Ferritin 1580

Steroid taper started, working diagnosis of CPPD and/or >Adult Onset Still's Disease. Patient discharged.

### Hospitalization #2

Recurrent sinus infections for next 6 months then returns with anterior uveitis.

### Notable Labs & Imaging:

#### Hematology:

WBC: nl (normal Eos) Hgb: 9.5 Plt: nl MCV: 102 (on B12 supp)

#### Chemistry:

CMP nl ESR high, Ferritin 1200, ACE 160 (elevated), 1-25 VD nl

Syphilis, Lyme, EBV, HIV, all negative. Ig levels not checked. ANCA neg, UDS negative

#### Imaging:

CTAP: mild hepatosplenomegaly without hilar/mediastinal lymphadenopathy. Inferior pole renal cyst.

Each abnormality was worked up separately. Iron studies were done and transferrin sat was 100%. C282Y mutation was found and presumed dx of hemochromatosis, but PLT count dropped suddenly to 22K. Thrombocytopenia workup negative.

PET: Multiple lytic lesions in long bones and pelvis. Increased uptake in lesions. Liver and spleen were hypermetabolic.

IG: IgA 0, IgM 0, IgG 470 Serum Kappa/Lambda ratio abnormal

EGD: diverticulosis

Bone marrow Bx: 95% replaced by plasma cells, kappa restricted, flow cytometry positive. Diagnostic of multiple myeloma, Congo red negative.

17 year old cousin had bilateral spontaneous AVN of knees. This was suspicious for Gaucher's Disease. Genetic and enzyme testing was sent for Gaucher's Disease and positive.

**Dx Gaucher's Disease**