A Delayed Diagnosis: Adult Onset Still’s Disease

Jacob N. DeLalla BS1, Nichole Brunton DO2, Rahila Ogunnaie MD 2
1American University of the Caribbean School of Medicine; 2Department of Internal Medicine, Danbury Hospital

Introduction

Objective: To highlight the diagnostic difficulties associated with Adult Onset Still’s Disease (AOSD), and clinical confounders that cause this to be an elusive diagnosis.

- AOSD is a systemic inflammatory disease presenting as a quotidian fever, evolving into an arthralgia and rash.
- Many cases of AOSD go unrecognized; approximately one third of cases self-resolve. Intermittent presentations may be frequently confused with other rheumatologic syndromes due to lack of definitive diagnostic testing.

Case Presentation

1. History of Present Illness:
   - 41-year-old Caucasian male
   - CC: Fever for 1 day, hematuria of 2 days (resolved), and improving ‘gouty’ bilateral knee and elbow joint pain ongoing for one year
   - PMH: Gout, & uric acid nephrolithiasis
   - Home medications: Prednisone as needed for joint pain, taking approximately 3 x per week, last taken on day of admission, and Fexofenadine 80 mg daily

2. Physical Examination:
   - Tachycardia.
   - Feburle, Tmx 102.4F
   - Minimal swelling in elbow, ankles and knees, otherwise unremarkable examination

3. Diagnostic Laboratory Results:
   - Ferritin = 3,875
   - CRP = 356.1
   - Ferritin >500 ng/mL

Initially admitted with a working diagnosis of pyelonephritis on broad spectrum antibiotics prior to admitting details about joint pains or rash.

Diagnostic Criteria for AOSD

Yamaguchi Diagnostic Criteria
5 or more criteria, at least 2 Major
- Major:
  1. Fever >39 C for >7 days.
  2. Arthralgia >2 weeks.
  3. Leukocytosis, >80% granulocytes.
- Minor:
  1. Sore throat
  2. Lymphadenopathy
  3. Hepatomegaly / Splenomegaly
  4. Abnormal liver function studies.
  5. Negative antinuclear antibody & rheumatoid factor.

Hemophagocytosis in bone
- Ferritin >500 ng/mL
- Ferritin >500 ng/mL

Diagnostic Laboratory Results:
- CRP = 356.1
- Ferritin = 3,875
- Triglycerides = 389

Pathology & Further Investigations

Hospital Day #4, new onset maculopapular rash over the trunk with elbow joint swelling, progressive anemia and high fevers:

1. Further Laboratory Results:
   - CRP = 356.1
   - Ferritin = 3,875
   - Triglycerides = 389
   - Mild transaminitis.

2. Imaging:
   - CT Abdomen/pelvis: retroperitoneal lymphadenopathy, splenomegaly, & nonspecific perinephric fat stranding.

3. Infectious disease & rheumatology consultations.
   1. Further history obtained revealed history of similar arthralgia & rash that improved after Prednisone use.
   2. Negative infectious work-up

Defining the Diagnoses

- Patient started on high dose steroid therapy after negative infectious work up
- Rash and joint pain improved in 24 hours
- Thrombocytosis occurred and anemia resolved in 72 hours

Given overlap of symptoms between AOSD and HLH (highlighted in the bottom middle), work-up initiated to rule out HLH as alternative etiology in the outpatient setting.

- Several cases of rheumatologic conditions, such as AOSD, triggering HLH have been reported.
- Negative outpatient work-up for malignancy suggests patient presented with a hybrid of both syndromes.

Conclusions

- A thorough patient history should always be obtained.
- Patients utilizing steroids on an ‘as needed’ basis should be closely followed to ensure appropriate use.
- This case highlights a unique presentation of two prognostically dissimilar pathologies (AOSD and HLH) sharing a significant diagnostic overlap.
- There is a need for improved diagnostic modalities for AOSD to minimize long-term adverse consequences and patient quality of life.

References