23. ADULT-ONSET STILL’S DISEASE PRESENTING AS SLE
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Introduction: A case of adult-onset Still’s disease initially diagnosed as lupus. A patient presented with clinical and immunological features consistent with SLE, however did not respond to conventional treatment. Over time she had recurrent flares associated with high inflammatory markers. Following extensive investigation she was diagnosed with adult-onset Still’s disease. She remains well controlled on tocilizumab and methotrexate. This was significant, as the history and results of initial investigation were consistent with SLE. Adult-onset Still’s disease was a diagnosis of exclusion following years of investigation. Diagnosis and an appropriate treatment regimen has resulted in prevention of further flares.

Case description: A 49-year-old lady presented in 2011 with a history of joint pain, rash and fevers. Her inflammatory markers were raised, CRP of 267.7 mg/L, white cell count 14.8 x 10⁹/L and neutrophil count 13.6 x 10⁹/L, ANA positive, with positive anti-RNP and anti-SM, dsDNA 73.3 IU/mL, pANCA positive, MPO 64. She was initially diagnosed with lupus. She was treated with multiple steroid sparing agents including mycophenolate mofetil, azathioprine and cyclophosphamide between 2011 and 2013. During this period she developed presumed cyclophosphamide-induced cardiomyopathy, and the cyclophosphamide was subsequently stopped. She has had recurrent flares presenting with rash and joint pain associated with raised inflammatory markers, neutrophilia and elevated ferritin, responding well to treatment with high dose steroids. During this time she has been extensively investigated for pyrexia of unknown origin. These investigations include two prior CT-PET scans in 2013 and 2015, which identified no source of sepsis or malignancy. Bone marrow aspirate performed 2015 demonstrated non-specific appearances with no malignancy. Symptomatic control was achieved for some time with methotrexate and low-dose steroids; however she had a significant flare and became acutely unwell with a rash, fever and raised inflammatory markers, requiring admission to intensive care in 2017. During this flare, her white cell count was 28.1 x 10⁹/L, neutrophil count 26.4 x 10⁹/L, ferritin was 11741 ug/L. She
responded well to pulsed methylprednisolone and made a good recovery. She was subsequently commenced on tocilizumab.

Following this episode, she was seen at the Periodic Fever Clinic at the Royal Free, where they confirmed a diagnosis of phenotypical adult-onset Still’s disease, however her genetic testing was negative. In the interim she has remained stable with well controlled symptoms on methotrexate and tocilizumab. Her most recent ANA and ENA have been negative, with normal inflammatory markers and ferritin since commencing current treatment.

Discussion: Initially due to a typical history of joint pain, rashes and fevers with an associated positive ANA, anti-RNP, anti-SM, raised dsDNA and positive pANCA, a diagnosis of SLE seemed highly likely. As time progressed, several different therapeutic agents were trialled, with varying success. She responded well to steroids, however steroid sparing agents did not seem to control her symptoms and she developed presumed cyclophosphamide induced cardiomyopathy, which led to this being withdrawn. She continued to have recurrent flares during this period. Alternative differential diagnoses were explored, including malignancy or unusual infections.

This was an interesting case, as this patient initially presented with a clinical and immunological phenotype typical of SLE. Due to the non-specific nature of adult-onset Still’s disease and its rarity, this was a diagnosis of exclusion. The persistent pyrexia with neutrophilia and raised ferritin during flares, and poor response to azathioprine, mycophenolate and cyclophosphamide were features more consistent with adult-onset Still’s disease, however she was ANA, RNP and SM positive until 2017, and remains pANCA positive.

Despite a phenotypical diagnosis of adult-onset Still’s disease and good response to treatment with methotrexate and tocilizumab, this patient’s genetic screening was negative. Her persistent positive pANCA and initial positive anti-nuclear antibodies are unusual for a patient with adult-onset Still’s disease, however the resolution of positive ANA, neutrophilia and raised ferritin with tocilizumab and methotrexate suggest an IL-6 mediated process.

Key learning points: This case was highly unusual and interesting, and highlighted several important points. Firstly, with many rheumatological conditions diagnosis is not always clear cut, and while this patient met the diagnostic criteria for SLE, she continued to have atypical flares and poor response to conventional steroid sparing agents. Differential diagnoses such as malignancy or infection have to be thoroughly investigated prior to diagnosis and commencing treatment for autoinflammatory conditions such as adult-onset Still’s disease. On a practical level, it can be challenging in today’s NHS to organise specialist investigations rapidly, especially as an outpatient.

This case has emphasised the importance of consideration of a wider differential for patients presenting with unexplained symptoms, especially when there is a poor response to conventional treatment.

Conflicts of interest: The authors have declared no conflicts of interest.