Autoinflammation and Myelofibrosis: Report of a Case

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Adult-onset Still’s disease (AOSD) is a rare systemic autoinflammatory disease of unknown etiology [1,2]. We describe the case of a young woman with AOSD presenting with extreme anemia and diagnosed with secondary myelofibrosis. Treatment with prednisolone resulted in durable complete clinical and hematologic response. She remains in excellent health, with normal blood counts during her four-year follow-up.

Case: A 17-yr-old girl was admitted to Mahavir Hospital in October 2018 with fever, pain in the knees, ankles, elbows and wrists, and shortness of breath. She denied small joint arthralgia or photosensitivity. She was admitted elsewhere in March 2018 with a history of fevers, pain in the knees and elbows, and severe anemia. She was transfused with packed erythrocytes and discharged on analgesics, oral iron and folic acid. She was hospitalized again in May, July and August of 2018 with similar complaints and required transfusion support. She was well between the flares of joint pains and fever. A bone marrow aspirate in August 2018 resulted in a dry tap; a trephine biopsy was not done.

On admission to Mahavir Hospital her temperature was 102° F, pulse 134 and respirations 24/min. There was no skin rash, lymphadenopathy or organomegaly. Her Hb was 1.3 g/dl, MCV 82 fl, reticulocytes 0.2%; leucocytes and platelets were normal. Blood smear examination showed normal red cell morphology. Results of routine biochemical tests including serum total and direct bilirubin and lactate dehydrogenase were normal. Ultrasound abdomen showed mild splenomegaly. Serum ferritin was 4084 ng/ml.
Results of direct antiglobulin test, antinuclear antibodies, and rheumatoid factor were negative. Bone marrow aspiration resulted in a dry-tap; trephine biopsy showed hypercellular marrow, normal trilineage hematopoiesis, scattered non-paratrabecular aggregates of lymphocytes, and MF-1 grade reticulin fibrosis (Figure 1). Tests for JAK2 V617F and CALR mutations were negative. Patient was transfused with four units of packed erythrocytes and discharged home on oral prednisolone 40 mg daily. Eight-weeks later, she was asymptomatic and her Hb was 12.3 g/dl; she had stopped taking prednisolone on her own ten days earlier. She remains well on no medications and her Hb was 13.4 g/dl in October 2022.

The findings in our patient satisfy the Yamaguchi criteria [3] for the diagnosis of AOSD. Its rarity and lack of diagnostic biomarkers often result in a delay in diagnosis of AOSD [2]. The marrow biopsy findings, normal red cell morphology, exclusion of alternate causes of myelofibrosis, and response to corticosteroid therapy are consistent with a diagnosis of autoimmune myelofibrosis in our patient. However, the formal diagnostic criteria of autoimmune myelofibrosis include the absence of preexisting “well-defined” autoimmune disease or autoimmune markers [4,5]. Our patient is considered to have autoinflammation-related myelofibrosis. We are not aware of prior reports of secondary myelofibrosis in AOSD. Whether the condition is underrecognized is not clear [2,6-8].

The occurrence of myelofibrosis in AOSD offers a bridge between autoimmunity and autoinflammation. A study of bone marrow reticulin in AOSD and other autoinflammatory diseases may help in understanding the role of systemic inflammation in bone marrow fibrosis.

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References