Systemic lupus erythematosus presenting to haematology with pancytopenia and features of macrophage activation syndrome

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DESCRIPTION

This bone marrow biopsy (figure 1) shows haemophagocytosis consistent with macrophage activation syndrome (MAS) secondary to previously undiagnosed systemic lupus erythematosus (SLE).

A 44-year-old woman had been unwell for 4 weeks with fever, weight loss and an aphthous ulcer. There were no other clinical features of SLE.

There was pancytopenia (platelet count 65×10^9/L; neutrophil count 0.5×10^9/L; haemoglobin 107 g/L). The reticulocyte count was 20×10^9/L. Parvovirus and Epstein-Barr virus IgM were not detected.

A very high ferritin level of 3717 µg/L in the context of cytopenias was suggestive of MAS, a life-threatening hyperinflammatory state.[1] Clinical features of MAS include fever, lymphadenopathy and hepatosplenomegaly. Laboratory markers include pancytopenia, altered liver function and coagulopathy, with raised lactate dehydrogenase (LDH) and triglycerides.[2]

This patient had raised aspartate transaminase and alanine transaminase levels (154 and 145 U/L, respectively). The LDH level was high (458 U/L). The triglyceride level was 2.07 mmol/L. There was no coagulopathy. The C-reactive protein level was 3.2 mg/L. There was a raised urine protein to creatinine ratio (>200).

Antinuclear antibody titres were raised (1/160–640). The anti-double-stranded DNA level was high at >379 IU/mL, which alongside ulceration, cytopenias and proteinuria yielded a diagnosis of SLE. Complement levels were low (C3 0.3 g/L and C4 0.1 g/L), indicating active disease. The cytopenias and clinical features responded well to prednisolone.

While the cytopenias may have been autoimmune in aetiology, the high ferritin level and bone marrow biopsy findings make MAS a likely contributing factor.

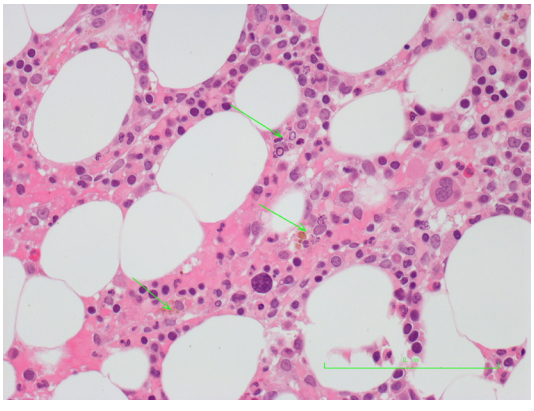

Figure 1 Bone marrow biopsy with abundant cell-bound haemosiderin and focal haemophagocytosis with cellular debris identified in macrophage cytoplasm (arrows). This is consistent with, but not specific for, macrophage activation syndrome.

Patient's perspective

"My SLE does affect my everyday life but I always think positively and feel very lucky – I feel better since the diagnosis."

"My hair is falling out which is depressing and I have to clean my room three times a week. Sometimes I have to do tasks slowly. Hot weather and stress makes it worse- my left ankle becomes swollen and I have an awareness of the joints in my toes. Initially I was not able to open bottles or peel onions but my joints are better now."

"I also bruise easily with steroids."

Learning points

- Uncommonly, systemic lupus erythematosus and other autoimmune or rheumatological disorders can present with macrophage activation syndrome (MAS), a life-threatening hyperinflammatory state where there is a constellation of fever with pancytopenia, with high ferritin, aspartate transaminase and triglyceride levels. The fibrinogen level is low.
- A very high serum ferritin level should trigger MAS as a differential, especially if the C-reactive protein level is normal or modestly elevated.
- Cytopenias with MAS secondary to autoimmune disease can respond to steroids alone, as in this case, as opposed to requiring chemotherapy.[3]

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