Macrophage activation syndrome in spondyloarthritis and monoallelic missense mutations in PRF1: a description of one paediatric case

Sirs,

Macrophage activation syndrome (MAS) is a serious, potentially fatal complication of paediatric rheumatic diseases, which is seen most commonly in systemic juvenile idiopathic arthritis (sJIA) (1). We report the case of a girl affected by B27-positive Spondyloarthritis (SpA) who developed recurrent episodes of MAS. The genetic analysis revealed the presence of a monoallelic missense mutation in the perforin gene (PRF1).

A 16-year-old girl was referred to us because of 2-week fever, unresponsive to oral antibiotics, dysuria, urticarial rash and buttock pain. The history until the age of 5 years as the familiar history are unknown because she was adopted. At the age of 11, she presented fever for 2 weeks with arthralgia and rash. In few days she developed a MAS according to the current criteria (2), characterised by urticarial rash, hepatosplenomegaly, hyperferritinaemia, hypertransaminasaemia, hypertrigliceridaemia, hyponatraemia, decrease in platelet count, confirmed by the presence of macrophage haemophagocytosis on bone marrow aspirate. The patient was successfully treated with steroids (3 pulses of methylprednisolone 30 mg/kg/day followed by prednisone 2 mg/kg/day), intravenous immunoglobulins (IVIG) 2 g/kg/day per 3 days and cyclosporine 4 mg/kg/day, progressively tapered and discontinued in 2 years.

After 6 months the patient presented a second episode of MAS and she was treated with steroids and cyclosporine achieving disease remission. Cyclosporine was discontinued 9 months before the present hospitalisation. At presentation, the patient had fever, dysuria, urticarial rash, buttock pain on the right side, psoriatic lesions on the forehead and acute bilateral uveitis.

Pelvic magnetic resonance imaging (MRI) disclosed right sacroiliitis (Fig. 1). HLA analysis revealed positive HLA-B27.

According to the current International League of Associations for Rheumatology (ILAR) classification of JIA (3), the patient was classified in the group of undifferentiated arthritis. Blood and urine cultures, collected in course of oral antibiotics, were negative, but the high phase reagents associated with macrophage activation syndrome in systemic juvenile idiopathic arthritis: diagnosis, genetics, pathophysiology and treatment. Genes Immun 2012; 13: 289-98.


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References:

Fig. 1. Magnetic resonance image. Coronal/Axial TSE images show diffuse perivascular bone marrow oedema of the right sacroiliac joint, consisting of sacroilitis.


