Recurrent infections and multiple bone fractures in a 4-year-old child

Ashok Vaghjimal, Jenny Shliozberg, Arye Rubinstein

A 4-year-old boy was referred to our clinic for evaluation of repeated skin infections, pneumonias, and recurrent otitis media starting from 9 months of age. The child had a history of eczema from one week of age. At the age of 3.5 years, he had sustained three fractures of long bones after a minor trauma. The family history was unremarkable. On physical examination he had an unusual and typical facies (figure), onychomycosis of finger nails and fading eczema on the right forearm. The rest of the physical examination was normal. The white cell count was elevated at 20.0 x 10^9/l with 20% eosinophils. Serum IgG, A and M were normal. T and B cells were also within normal limits, both as percentages and in absolute numbers. Delayed-type hypersensitivity skin tests with Candida, tetanus toxoid and purified protein derivative were negative.

Questions

1. What tests should be performed?
2. What is the probable diagnosis?

Albert Einstein
College of Medicine,
Division of Allergy and Immunology, Mazer
Building Room 200,
1300 Morris Park Avenue, Bronx, NY
10461, USA
A Vaghjimal
J Shliozberg
A Rubinstein

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Answers

QUESTION 1
Serum IgE should be measured and a leucocyte chemotaxis test performed. Serum IgE was 23 000 IU/ml in our patient (normal range 8–69 IU/ml). The leucocyte chemotaxis test was normal.

QUESTION 2
Hyper-IgE syndrome (Job’s syndrome).

Discussion

Originally described in two red-haired girls with recurrent ‘cold abscesses’, Job’s syndrome is a rare disorder characterised by elevated serum IgE levels, eczema and recurrent bacterial infections (box 1). The eczema is atypical and is present in nearly all patients. The affected skin is prone to bacterial and viral super-infections.

Recurrent pneumonias are the most serious infections in Job’s syndrome, often resulting in pneumatocele formation and empyema. Oral thrush, gingivitis and onychomycosis are described less frequently. Coarse facial features with a broad nasal bridge are present in the majority of the patients. Osteoporosis has also been reported in some patients and may be associated with an increased incidence of bone fractures.

A number of immunological abnormalities have been described in Job’s syndrome; the main ones are summarised in box 2. The white cell count may also be elevated, as was the case in our patient.

No specific therapy exists for this syndrome. Early recognition of infections and prompt treatment is necessary to prevent various complications. Dicloxacillin and trimethoprim–sulphamethoxazole have been used successfully to prevent infections. Topical steroids, emollients and antihistamines should be used to treat atopic dermatitis. Pneumococcal, influenza and varicella vaccines are also recommended, although multiple boosts may be necessary with the pneumococcal vaccine to obtain adequate antibody titres. Experimental therapies of Job’s syndrome include plasmapheresis and intravenous immunoglobulin.

Final diagnosis

Hyper-IgE syndrome (Job’s syndrome).

Keywords: hyperimmunoglobulin E; Job’s syndrome

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