

Case Report

An uncommon presentation of pneumonic Koch's with various splenic ulcer

Priyanka V. Kareena

Department of Clinical Microbiology and Immunology, Faculty of Medical Laboratory Science, University of Mumbai, Mumbai, India. Email: prof.kareena22@yahoo.co.uk

Accepted 15 February, 2015

Job's syndrome is an immunodeficiency disorder with defect in leukocyte chemotaxis in some patients. It is also called hyper IgE recurrent infection syndrome. Major manifestations are pruritic dermatitis, recurrent Staphylococcal pneumonias, mucocutaneous candidiasis, restrictive lung disease, scoliosis with other various features. A 20 year old boy who was presented to us with heart failure, abdominal pain and pyoderma was later diagnosed as having job's syndrome and tuberculosis.

Key words: Job's syndrome, hyper immunoglobulin E syndrome, Buckley syndrome, immunodeficiency.

INTRODUCTION

Job's syndrome, which was first described in 1966 (Davis et al., 1966), and which was also called hyper immunoglobulin E syndrome or Buckley syndrome, is a rare immunodeficiency disorder with Autosomal Dominant inheritance, variable expressivity and is associated with multiple abnormalities. The most common finding are recurrent skin abscesses, hence the name job's syndrome. Others are pneumonia with pneumatocele development and increased serum levels of Immunoglobulin E (IgE). Facial, dental and skeletal features are also associated with this syndrome. Non-immunologic features are impaired shedding of primary teeth, recurrent bone fracture, hyper-extensible joints and scoliosis. Although most cases are sporadic, multiple families with AD and AR inheritance have been described. Some authorities believe 2 separate syndromes exist, not one. Type 1 Hyper-immunoglobulin E have abnormalities in multiple systems including skeletal, dental and immune system, whereas, Type 2 Hyper-immunoglobulin E syndrome shows abnormalities confined to immune system. Hypomorphic mutations have been found in the signal transducer and activator of transcription 3 (STAT 3) genes in Type 1 and null mutation in tyrosine kinase 2 Tyk 2 gene in type 2 (Bonillafa, 2006; Grimbacher et al., 2005).

CASE STUDY

A 20 years old boy from karauli district of Rajasthan, India (Figure 1), came with complaints of difficulty in

breathing, skin lesion in front of chest and abdominal pain with fever on and off occurring over 1 month. Difficulty in breathing was more in lying down than sitting or standing, increased in night and relieved by treatment (diuretics) taken at periphery. Abdominal pain was intermittent, moderate, relieved by simple analgesic. It was associated with sense of fullness in whole abdomen and decreased appetite. Numerous eczematous skin lesions (pyoderma) associated with itching in front of chest were also present. Not relieved by treatment taken at periphery. There was history of significant weight loss in this one month. No history of cough/night sweats. On examination there was moderate pallor, scoliosis towards the right at 30°, moderate hepato splenomegaly with tender spleen and bilateral extensive crepts in both lung fields especially basal. No lymphadenopathy or no sternal tenderness was present. Rest of gastrointestinal, cardiovascular and neurological examination was within normal limits. On anterior chest wall there were numerous eczematous lesions measuring 4 × 6 mm.

Laboratory investigations revealed microcytic anemia with hemoglobin 7.6 g/dl MCV-73.7fl, MCH-25.6 pg MCHC-34.7%, Platelets-2.49 lakhs/mm³, total leukocyte count-26000/mm³, serum lactate dehydrogenase was 1013 U/L, erythrocyte sedimentation rate was 100. Peripheral smear showed no parasites. Differential leukocyte count showed eosinophils (5%), basophils (0.0%), neutrophils (89.9%) and Lymphocytes (6.9%). Renal functions, liver functions along with urine routine were normal. Serum albumin was 2.4 g/dl, serum globulin 5.1 g/dl, total proteins 7.5 g/dl. Test for enteric



Figure 1. Patient with jobs syndrome with skin lesions.

and malaria were negative. Chest x ray showed miliary mottling of both lung fields. Ultrasound abdomen showed multiple splenic abscesses with the largest as 3 × 4 cm and HIV1/2 was negative. Sputum for acid fast bacilli was negative. Montaux test was also negative. Polymerase chain reaction (PCR) for tuberculosis was positive. IgE level was 2496.50 IU/ml [NORMAL= 0 - 158.0 iu/ml]. Immunoglobulin G (IgG) level was 2419.0 mgs/dl [N=700-1600 mg/dl]. Immunoglobulin M (IgM) level 100.0 mgs/dl [N=40-230]. Immunoglobulin A (IgA) level 666 iu/ml [N=70-400]. So IgA, E and G levels were increased and IgM was normal. Polymerase chain reaction based on high resolution DNA melting assay for STAT 3 was positive. Neutrophil chemotaxis by sodium caseinate was abnormal. The patient was transfused with 3 units of packed cells and was started on cat 1 ATT with Intravenous antibiotics including linezolid, metronidazole, amikacin and 3rd generation cephalosporin for skin lesion, the skin department suggested antihistaminics orally and steroid creams locally. For splenic abscess, splenectomy was done by surgeons due to septicemia and immunization given as protocol. Biopsy showed staphylococci to be the cause. Patient discharged after 15 days postoperatively when he was stable and in a

good condition. The follow-up that was done after 1 month showed that the patient was in good condition.

DISCUSSION

Job's syndrome as previously described as a multiple system primary immunodeficiency syndrome. It is a rare disorder; about 250 to 300 cases have been published. The mortality of job's syndrome is mainly because of severe systemic infections. It occurs in all ethnic groups with no sex predilection. Diagnosis is based on increased IgE levels with focuses of infections like splenic abscess, recurrent bronchitis, pneumonia especially *Staphylococcus aureus* or *Haemophilus influenzae*. Coronary artery aneurysms have also been described. Others may have otitis externa, sinusitis, caries, and gingivitis or cervicofacial infections. IgE levels are usually >2000 IU/ml, many have >5000 IU/ml, eosinophils may be variable increased/normal, but do not correlate with disease activity. A normal IgE cannot exclude Job's in an adult. In some patients, disease activity decreases over years; IgE can decrease by effective and early treatment, patient's can lead a productive life.

Prophylaxis with penicillin /those resistant to penicillins, cephalosporins is recommended. Treatment at present in Indian subcontinent is symptomatic and supportive with focus on infections, and educating different families about the disease.

MATTER OF CONFLICT

Our patient's showed elevations of IgG, IgA and IgE. Both IgA and IgE increased in Wiskot Aldrich syndrome, however, IgM decreased in Wiskot, which was not in our case. Elevation of IgG and IgA can be explained in part by chronic infection most likely to be Koch's chest in our patient (Anthony et al., 2008).

REFERENCES

- Anthony S Fauci, Braunwald E, Kasper DL, Hauser SL, Longo DL, Jameson JL, Loscalzo J (2008). Harrison's Principles of Internal Medicine 17th edition; USA; McGraw-Hill Companies, Inc., p. 381.
- Bonillafa GRs (2006). Update on primary immunodeficiency disorder. *J. Allergy Clin. Immunol.*, 117: 4535.
- Davis S, Schaller J, Wedgwood R (1966). Job's Syndrome. Recurrent, cold, staphylococcal abscesses. *Lancet*, 1(7445): 1013-1015.
- Grimbacher B, Holland SM, Puck JM (2005). Hyper IgE Syndromes. Department Rheumatology Clinical Immunology, Medical SCHOOL, University of Freiburg, Germany, 203: 244-250.