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Hyper-Immunoglobulin E Syndrome in a Neonate: A Case Report

Bir yenidoğanda Hiperimmunglobulin E sendromu: Olgu sunumu

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Abstract- Hyper-immunoglobulin E syndrome (Job syndrome) is a rare primary immunodeficiency with variable presentation, characterized by recurrent infections, facial dimorphism, eczema, scoliosis, joint hyper-extensibility, pathologic fractures, very high IgE (>2000 IU/mL), severe eosinophilia and variable impaired T cell function. We present a case of Hyperimmunoglobulin E syndrome in neonate with review of the literature. *J Microbiol Infect Dis* 2013; 3(3): 144-146.

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Özet- Hiper-immunglobulin E sendromu (Job Sendromu), genellikle çok yüksek IgE (> 2000 IU/ml) seviyesi, şiddetli eozinofili, soğuk stafilokok cilt absesi ve pnömoni gibi tekrarlayan enfeksiyonlar, egzama, skolyoz, eklem hiperextansibilitesi, patolojik kırıklar, tipik bir yüz görünümü, kraniyosinostoz ve değişken bozulmuş T hücre fonksiyonu ile karakterize nadir görülen primer κ ubclass yetmezlik durumudur. Hiper-immunglobulin E sendromu yenidoğan ve diğer yaş grubunda farklı laboratuvar bulguları, klinik belirti ve bulguları gösterebilir. Bu çalışmada soğuk Stafilokokal cilt abseleri, hafif yüksek total serum IgE düzeyi (146 IU/ml, normal: 0-8 IU/ml), yüksek κ ubclass eozinofili (% 15) ve normal serum IgA, IgG, IgG κ ubclass, IgM, C3 ve C4 seviyelerine sahip hiper-immunglobulin E sendromu olan onbeş günlük erkek hasta sunuldu.

Anahtar Kelimeler: hiper-immunglobulin E sendromu, job sendromu, yenidoğan.

I. INTRODUCTION

Hyper-immunoglobulin (Ig) E syndrome (HIES or Job Syndrome) is a rare primary immunodeficiency generally characterized by recurrent infections such as staphylococcal cold skin abscesses and pneumonia, eczema, scoliosis, joint hyperextensibility, pathologic fractures, a typical facial appearance, craniosynostosis, very high IgE, severe eosinophilia, and variable impaired T cell functions. The mechanisms responsible for hyperproduction of IgE and eosinophils in patients with HIES are presently unknown. Generally the onset of HIES occurs in children and elderly individuals.^{1,2} HIES may have variable presentation, and laboratory values in different age groups.^{3,4}

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We present a 15-days old newborn with HIES whose only have staphylococcal cold skin abscesses eosinophilia and high immunoglobulin E levels.

II. CASE REPORT

A fifteen-days-old male neonate born at 40 weeks of gestation by normal spontaneous vaginal birth to a 24 years-old mother without history of significant disease such as eczema or HIES in the family. The antenatal ultrasonography was normal. The patient was admitted to Dicle University Hospital at fifteenth day of his life, because of cold abscess that appeared 5 days before admission. On physical examination there was a 2x3 cm swelling compatible with cold abscesses in the anterior right knee area, right supraclavicular area, lateral right chest area and anterior left ankle area. He also had a characteristic facial appearance such a broad nasal bridge, cheilitis, thickened skin, and deep-set eyes with a prominent chin and forehead (Figure 1-3). There were no eczematous rash, scoliosis, fractures history, joint hyperextensibility and craniosynostosis on his physical examination and history. Yolbaş İ, et al. Hyper-immunglobulin E syndrome 145 *J Microbiol Infect Dis* www.jmidonline.org Vol 3, No 3, September 2013 The patient had slightly higher total serum IgE level (146 IU/mL, normal range: 0-8 IU/mL), high peripheral eosinophilia (15%) and normal serum IgA, IgG, IgG subclasses, IgM, C3 and C4 levels. The patient's other biochemical parameters were normal. The neonate underwent incision with pus aspirated which later grew *Staphylococcus aureus*. The skin biopsy showed eosinophil infiltration. Staphylococcal cold skin abscesses were treated with Ampicillin-sulbactam after drainage. HIES was diagnosed by clinically and laboratory tests, because there are no genetic or other confirmatory tests available in Turkey. The patient's computed tomography of the lungs was normal. The patient's Dual-energy X-ray absorptiometry test was found normal. The patient was discharged after two week from the hospital without any complications.

III. DISCUSSION

HIES is a multi-system disorder with a wide range of clinical phenotypes and signs, including skeletal, connective tissue, and vascular abnormalities.³ Most of patients with HIES suffer from recurrent staphylococcal infections of skin and lungs.⁴ Generally recurrent pyogenic pneumonias start in early childhood, and the most common infecting organisms are *Staphylococcal aureus*, *Haemophilus influenzae* and *Streptococcus pneumoniae*. Also mucocutaneous candidiasis is common in HIES.⁴ Musculoskeletal abnormalities of HIES are scoliosis, osteopenia, minimal trauma fractures, hyperextensibility and degenerative joint disease.^{3,5} The patients with HIES may have problem with development of their teeth.⁶ Our case had multiple cold skin abscesses in the various regions of body but had no other stigmata of HIES at this age. Characteristic facial appearance of HIES include broad nasal bridge, cheilitis, thickened skin, and deep-set eyes with a prominent chin and forehead.³ Our case had the similar characteristic facial appearance such as broad nasal bridge, cheilitis, thickened skin, and deep-set eyes with a prominent chin and forehead.

The two most consistent laboratory abnormalities in HIES are eosinophilia and elevated serum IgE. Over time, the serum IgE may decline in adults or may increase in newborn.³ Demirci et al⁷ found that IgE level of a two-month-old patient with HIES was 75.3 IU/ml (Range: 15-32 IU/ml), But in the same patients' they found IgE level 13,000 IU/ml after eight months. The patients with HIES have normal serum IgM, IgG, and IgA levels.³ Our case have had slightly higher total serum IgE level as 146 IU/ 146 Yolbaş İ, et al. Hyper-immunglobulin E syndrome J Microbiol Infect Dis www.jmidonline.org Vol 3, No 3, September 2013 mL (Normal range: 0-8 IU/mL) and high peripheral eosinophilia (15%).

The diagnosis of HIES is usually made based on characteristic facial appearance and clinical features associated with high serum IgE level and eosinophilia.⁵ Our patient had some of the characteristic features and laboratory findings. However definitive diagnosis is made on genetic basis such as STAT3.

Management of HIES currently revolve around prevention and treatment of infections. There is no cure for HIES at present. Therapy includes drainage of cutaneous abscesses followed by intravenous antibiotic therapy directed against mostly staphylococcal aureus. Prophylactic antibiotics and specific treatment is based on organ involvement. Immunoglobulin replacement therapy and some other treatments such as IFN-g, IFN-a, histamine-2 antagonists, and cyclosporine have been tried, which seem to be useful in the management of patients with HIES.^{2,8} Prophylactic antibiotic or antifungal prophylaxis (e.g., trimethoprim-sulfamethoxazole or fluconazole) should be recommended in the patients



Figure 1 : Characteristic facial appearance such a broad nasal bridge, cheilitis, thickened skin, and deep-set eyes with a prominent chin and forehead, and Staphylococcal cold skin abscesses on right supraclavicular area



Figure 2 : Staphylococcal cold skin abscesses on lateral right chest area



Figure 3 : Staphylococcal cold skin abscesses on anterior

with HIES with recurrent sinopulmonary, cutaneous infections, mucocutaneous candidiasis and invasive fungal infections.⁸

In conclusion, HIES may present with some features in the newborn baby. Recognition of leading signs of the disease will provide early diagnosis and prophylactic measures.

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