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Case Study

A Rare Case of Hyper-IgE Syndrome in a Two-Year-Old Child with Recurrent Pneumonia and Generalized Eczematous Dermatitis

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ABSTRACT

Hyper-IgE syndrome (HIES), also known as Job syndrome, is a rare primary immunodeficiency disorder characterized by markedly elevated serum immunoglobulin E (IgE) levels, recurrent sinopulmonary and cutaneous infections, chronic eczematous dermatitis, eosinophilia, and skeletal or connective tissue abnormalities. The autosomal dominant form is most commonly caused by mutations in the STAT3 gene, resulting in impaired T-helper 17 (Th17) cell differentiation and defective neutrophil recruitment. The rarity of the disorder, overlapping clinical manifestations with atopic dermatitis and bronchial asthma, and variable disease severity frequently delay diagnosis, thereby increasing the risk of irreversible pulmonary complications such as bronchiectasis and pneumatoceles. Early recognition and multidisciplinary management are therefore essential to reduce morbidity and improve long-term outcomes.

INTRODUCTION

Hyper-IgE syndrome (HIES), also known as Job syndrome, is a rare primary immunodeficiency characterized by markedly elevated serum immunoglobulin E (IgE) levels, chronic eczematous dermatitis, recurrent skin and respiratory tract infections, eosinophilia, and variable skeletal and connective tissue abnormalities. The syndrome was first described by Davis et al. in 1966 in patients who developed recurrent "cold" staphylococcal abscesses, resembling the suffering of the biblical figure Job. Later, Buckley and colleagues identified markedly elevated serum IgE levels in association with eczema and recurrent infections, leading to the recognition of Hyper-IgE syndrome as a distinct clinical entity.[1,2]

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HIES is an extremely uncommon disorder, with an estimated prevalence of fewer than one case per million people. It affects both males and females and has been reported worldwide. The disease is genetically heterogeneous and is broadly classified into autosomal dominant (AD-HIES) and autosomal recessive (AR-HIES) forms. Most cases of AD-HIES result from mutations in the STAT3 gene, whereas AR-HIES has been linked to mutations in genes such as DOCK8, ZNF341, and ERBIN, each contributing to defects in immune regulation and host defense.[3,4]

The underlying immune dysfunction in HIES is primarily related to impaired STAT3 signaling, which disrupts the differentiation of T-helper 17 (Th17) cells. This defect reduces the production of interleukin-17 and interleukin-22, cytokines that play an important role in protecting the skin and mucosal surfaces against bacterial and fungal pathogens. As a result, affected individuals are highly susceptible to recurrent infections, particularly those caused by Staphylococcus aureus, Haemophilus influenzae, and Streptococcus pneumoniae. In addition to recurrent infections, abnormalities in connective tissue remodeling may lead to skeletal manifestations, delayed shedding of primary teeth, scoliosis, hyperextensible joints, and characteristic facial features in patients with autosomal dominant disease.[3–5]

The clinical manifestations of HIES usually begin during infancy or early childhood. Persistent eczema is often the earliest feature and may initially be mistaken for severe atopic dermatitis. Over time, patients frequently develop recurrent pneumonia, chronic otitis media, sinusitis, skin abscesses, mucocutaneous candidiasis, and recurrent wheezing. Repeated pulmonary infections can result in long-term complications such as bronchiectasis and pneumatoceles, which contribute significantly to disease-related morbidity. Laboratory findings typically include marked eosinophilia and extremely elevated serum IgE levels, although these findings should always be interpreted in conjunction with the patient's clinical presentation. Diagnosis is based on characteristic clinical features, immunological investigations, the National Institutes of Health (NIH) clinical scoring system, and, whenever possible, confirmation by genetic testing.[5,6]

Because many of its clinical features overlap with common allergic disorders such as atopic dermatitis and bronchial asthma, the diagnosis of HIES is often delayed, particularly in young children. Failure to recognize the condition early may allow recurrent infections to cause irreversible pulmonary damage and other systemic complications. Therefore, prompt identification of affected patients, early initiation of appropriate antimicrobial therapy, meticulous skin care, regular immunological evaluation, and multidisciplinary follow-up are essential to reduce complications and improve long-term outcomes.[4,7]

In this report, we describe the case of a two-year-old boy who presented with generalized pruritic dermatitis, recurrent pneumonia, bronchial asthma, eosinophilia, and markedly elevated serum IgE levels, consistent with Hyper-IgE syndrome. This case highlights the importance of considering primary immunodeficiency disorders in children with persistent eczema and recurrent respiratory infections, as early diagnosis and comprehensive management can significantly improve prognosis and quality of life.

CASE PRESENTATION

We report the case of a two-year-old male who presented with generalized pruritic erythematous skin lesions involving the face, trunk, upper limbs, and lower limbs. The child had a significant medical history of recurrent pneumonia, bronchial asthma, recurrent wheezing episodes, eosinophilic lung disease, and multiple hospital admissions

since infancy. Clinical examination revealed diffuse eczematous dermatitis without acute respiratory distress. Laboratory investigations demonstrated leukocytosis (20,720 cells/mm³), eosinophilia (32%), mild microcytic hypochromic anemia (hemoglobin 10.6 g/dL), elevated inflammatory markers, and biochemical parameters within normal limits. Previous investigations documented markedly elevated serum IgE levels with positive Aspergillus-specific IgE, supporting the diagnosis of Hyper-IgE syndrome in conjunction with the characteristic clinical phenotype.

The patient was managed with intravenous cefotaxime for suspected secondary bacterial infection, oral antihistamines, topical corticosteroids, emollient therapy, inhaled budesonide, bronchodilator therapy, and supportive skin care. Clinical improvement was observed during hospitalization with reduction in pruritus, gradual resolution of cutaneous lesions, and stabilization of respiratory symptoms. The child was discharged on maintenance therapy with instructions for regular pediatric immunology follow-up, infection surveillance, and long-term dermatological care

MATERIALS AND METHODS

This case report describes a 2-year-old male diagnosed with Hyper-IgE syndrome who presented with recurrent respiratory infections and generalized pruritic dermatitis. Clinical data, laboratory investigations, radiological findings, treatment details, and follow-up information were collected retrospectively from the patient's medical records. The diagnosis was based on characteristic clinical features, elevated serum IgE levels, eosinophilia, and recurrent infections. Patient confidentiality was maintained, and informed consent for publication was obtained from the patient's guardian

DISCUSSION

Hyper-IgE syndrome (HIES), or Job syndrome, is a rare primary immunodeficiency characterized by markedly elevated serum IgE levels, recurrent bacterial infections, chronic eczema, eosinophilia, and varying skeletal abnormalities. The disorder most commonly results from mutations in the STAT3 gene, leading to impaired T-helper 17 (Th17) cell differentiation and defective neutrophil recruitment, thereby increasing susceptibility to recurrent skin and respiratory infections.[3,4] The present case demonstrated the classical clinical features of HIES, including generalized pruritic dermatitis, recurrent pneumonia, bronchial asthma, eosinophilia, and elevated serum IgE levels, supporting the clinical diagnosis.

Eczema is often the earliest manifestation of HIES and is frequently misdiagnosed as severe atopic dermatitis. Freeman and Holland reported that recurrent eczema associated with repeated respiratory infections should prompt evaluation for an underlying primary immunodeficiency rather than isolated allergic disease.[3] Similar to previous reports, our patient presented with diffuse pruritic skin lesions accompanied by recurrent pulmonary infections, highlighting the importance of recognizing this characteristic clinical combination.

Pulmonary complications remain the major cause of morbidity in HIES. Holland et al. demonstrated that recurrent pneumonias caused by organisms such as Staphylococcus aureus and Streptococcus pneumoniae can progress to bronchiectasis and pneumatoceles if diagnosis and treatment are delayed.[4] Our patient had multiple episodes of pneumonia and wheezing since early childhood, emphasizing the need for early diagnosis and long-term respiratory follow-up to prevent irreversible lung damage.

Marked eosinophilia and elevated serum IgE are characteristic laboratory findings in HIES but should be interpreted alongside clinical manifestations. Minegishi emphasized that these immunological abnormalities, although not disease-specific, strongly support the diagnosis when associated with recurrent infections and eczema.[5] In our patient, eosinophilia and elevated IgE, together with recurrent respiratory infections and dermatitis, were highly suggestive of HIES. Management focused on controlling acute infection and skin inflammation using intravenous antibiotics, inhaled corticosteroids, bronchodilators, antihistamines, topical corticosteroids, and emollients, resulting in clinical improvement.

This case highlights the importance of considering Hyper-IgE syndrome in children presenting with persistent eczema, recurrent pneumonia, and eosinophilia. Early recognition, multidisciplinary management, and regular follow-up are essential to reduce infectious complications, prevent irreversible pulmonary damage, and improve long-term quality of life.[3,4,7]

CONCLUSION

Hyper-IgE syndrome is a rare but important primary immunodeficiency that should be suspected in children presenting with persistent eczema, recurrent respiratory infections, eosinophilia, and markedly elevated serum IgE levels. Early recognition and prompt multidisciplinary management are essential to reduce recurrent infections, prevent irreversible pulmonary complications, and improve long-term outcomes. This case highlights the importance of considering HIES in the differential diagnosis of severe atopic dermatitis with recurrent pneumonia, thereby facilitating timely diagnosis and appropriate therapeutic intervention

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