SCIENTIFIC FEDERATION
ABODE FOR RESEARCHERS

Fehmida Najmuddin, SF J AIDS HIV Res, 2017, 1:1

SciFed Journals of AIDS & HIV Research

Case Report Open Access

# Hyper-IgE Syndrome: A Rare Entity

[1]Fehmida Najmuddin, [2]Deepak Sharma, [3]Rajesh Rai, [4]Sadaf Siddiqui

*Dr D Y Patil Hospital,Medical College and Research Centre, Navi Mumbai*

**Keywords**

Hyper IgE Syndrome; Pediatrics; Recurrent Infections; Hypereosinophilia

## Introduction

HYPER IgE syndromes (HIS) are a group of rare immunodeficiency disorders involving the innate immunity. They are majorly divided into 2 forms; autosomal dominant (AD) and autosomal recessive (AR) with the former having a higher incidence of occurrence. The autosomal dominant form is due to mutation in the STAT-3 gene [1] while the autosomal recessive one is due to mutations in the gene encoding DOCK8 [2]. HIS is characterized by recurrent systemic infections with markedly elevated levels of serum IgE levels [3]. We report a case of 2 year old child with recurrent infections which was later diagnosed as HIS after the common causes were ruled out.

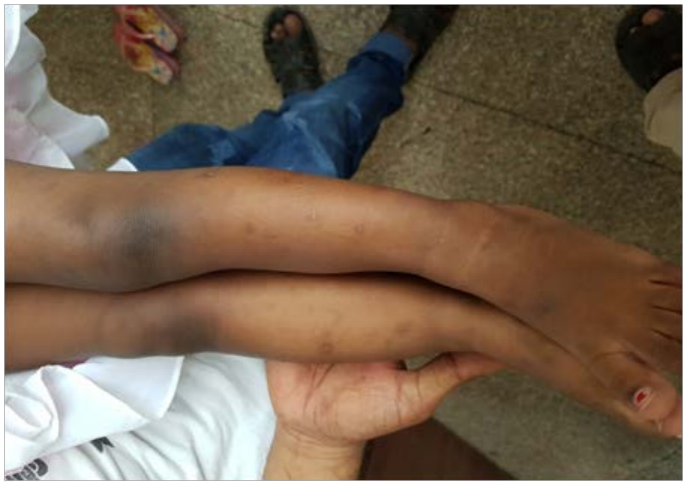
## Case Report

2 year old female child born of non-consanguineous marriage residing in Mumbai came to a tertiary care centre with complaints of cough since 5 days and breathlessness since 1 day. Cough was non spasmodic, non- productive associated with sudden onset progressive breathlessness with no diurnal or postural variation. There was no history of fever, foreign body aspiration, post – tussive vomiting or ear discharge.

Past History revealed significant hospital admissions for recurrent pneumonias, skin infections, Gastroenteritis and urinary tract infection since the last one year. There is no family history of "Atopy", allergy or asthma with Birth history being uneventful. Child was partially immunized and developmentally normal. They belonged to the lower socioeconomic strata of society.

On examination, child was a febrile, with HR- 120/min, RR- 54/min with respiratory distress. Anthropometric measurements revealed grade I protein energy malnutrition with no stunting. There was pallor, hyper pigmented, eczematous lesions over the upper and lower limbs with an abscess over the right nipple (Figure 1, Figure 2) and no coarse facial features. Respiratory examination showed reduced air entry, "Crepetitions" and rhonchi bilaterally with other systems being normal.

**Figure 1:** Eczematous lesions over the lower limbs

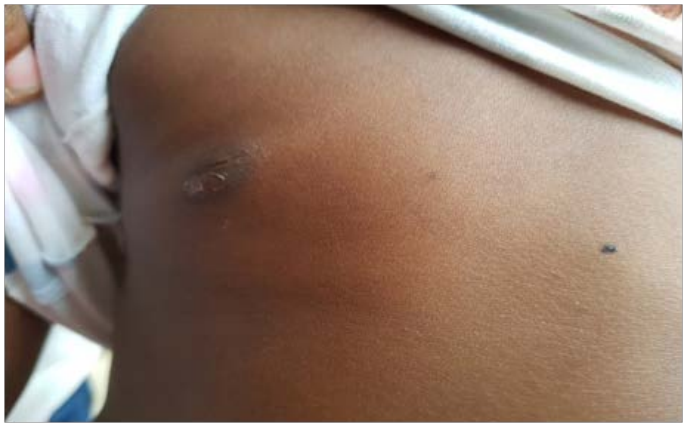


***Corresponding author:** Fehmida Najmuddin, Dr D Y Patil Hospital,Medical College and Research Centre, Navi Mumbai. **E-mail:** fehmidanc@hotmail.com. **Tel:** 0091-9920030533

**Received** October 03, 2017; **Accepted** October 11, 2017 ; **Published** October 25, 2017

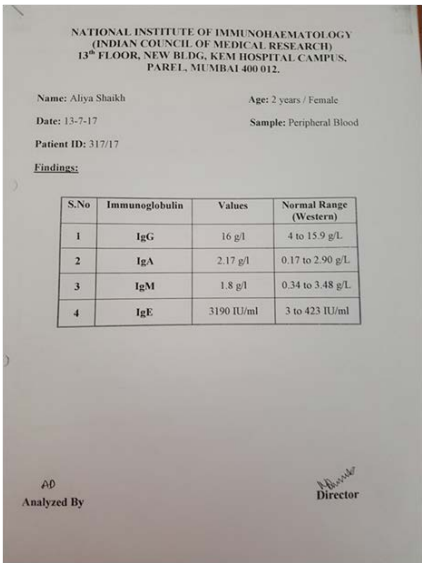
**Citation:** Fehmida Najmuddin (2017) Hyper-IgE Syndrome: A Rare Entity. SF J AIDS HIV Res 1:1.

**Figure 2:** Showing an abscess over the chest



On investigations, Complete blood count (CBC) revealed Hemoglobin – 8.1 gm/dl with microcytic hypo chromic anemia, Total Leucocytes Counts-18,600/µl, ( Neutrophils- 48.3%, Lymphocytes-40%, Eosinophils-8.7%, Monocytes-2.9% and Basophils-0.1%) with Hypereosinophilia and Absolute Eosinophilic count-1600/µL. Blood culture showed no growth and chest X Ray was suggestive of bronchopneumonia. Tuberculosis, Human immunodeficiency Virus (HIV) Tri-Dot test, stool routine and culture were negative. On the basis of significant recurrent hospital admissions and persistent eosinophilia, we suspected immunodeficiency syndromes and sent the blood samples for flow cytometric analysis, NBT (Nitro blue tetrazolium) slide test which was normal. The immunoglobulin assay revealed Increased IgE levels – 3190 IU/Ml with normal IgG, IgA, and IgM levels

**Figure 3:** Raised Serum IgE levels



NATIONAL INSTITUTE OF IMMUNOHAEMATOLOGY
(INDIAN COUNCIL OF MEDICAL RESEARCH)
13th FLOOR, NEW BLDG, KEM HOSPITAL CAMPUS,
PAREL, MUMBAI 400 012.

**Name:** Aliya Shaikh **Age:** 2 years / Female
**Date:** 13-7-17 **Sample:** Peripheral Blood
**Patient ID:** 317/17
**Findings:**

| S.No | Immunoglobulin | Values | Normal Range (Western) |
|---|---|---|---|
| 1 | IgG | 16 g/l | 4 to 15.9 g/L |
| 2 | IgA | 2.17 g/l | 0.17 to 2.90 g/L |
| 3 | IgM | 1.8 g/l | 0.34 to 3.48 g/L |
| 4 | IgE | 3190 IU/ml | 3 to 423 IU/ml |

AD
Analyzed By
Director

We started the child on broad spectrum antibiotics, anti-helminthic and supportive therapy, following which the child did not improve clinically and repeat CBC showed persistent eosinophilia. The recurrent skin lesions were diagnosed as atopy dermatitis and therapy for the same was initiated.

With the above clinical picture and investigations, patient was diagnosed as Hyper IgE syndrome, counseled for bone marrow transplantation and as in on regular follow up.

## Discussion

The autosomal dominant form of HIS is characterized by recurrent pneumonias, sinopulmonary and fungal infections as compared to the AR form which is more commonly associated with skin, food allergies and Asthma. AD forms are strongly associated with history of musculoskeletal involvement and coarse facies which is absent in the AR form [4, 5].

Since tuberculosis and Acquired Immunodeficiency syndrome (HIV) is endemic in our country, our initial line of investigation was to rule out the above causes. The child also had recurrent atopic dermatitis and pneumonias, which further compelled us to investigate for common allergic cascade of diseases and treatment was initiated for the same. Since the patient did not have any relief in symptomatology and had repeated episodes of pneumonia and skin infections in the form of areolar abscess, we suspected immunodeficiency disorder, and on investigations, diagnosed the child with Hyper IgE syndrome in view of very high levels of Serum IgE (> 2000 IU/ml).

Our patient was having clinical manifestations which were likely of an AR form of HIS. Genetic mutational analysis could not be done due to financial constraints.

The treatment of AD HIS is a long term administration of anti-staphylococcal antibiotics and Intravenous Immunoglobulin [6, 7]. The treatment of choice in AS HIS is allogenic bone marrow transplantation [8] and has a poorer prognosis as compared to the AD HIS.

Serum IgE levels can be increased in diseases like atop, churg Strauss syndrome, Olmsted syndrome, Wiskott - Aldrich syndrome, omen syndrome, nezeloff syndrome and anti-helminthic infections [9, 10].

With the above report we want to highlight the rarity of this syndrome, the need for a high index of suspicion particularly in those children who have allergic symptoms and don't respond to the therapy with recurrent

pneumonias. Lastly though high serum IgE levels are associated with allergic cascade, one should always rule out Hyper IgE syndromes if the levels are above 2000 IU/ml with characteristic clinical features.

**References**

1. Yong PF, Freeman AF, Engelhard KR, et al. (2012) An update on the hyper IgE syndromes. Arthritis Res Ther 14: 228-238.

2. Engelhard KR, McGhee S, Winkler S, et al.(2009) Large deletions and point mutations involving the dedicator of cytokinesis 8(DOCK8) in the autosomal recessive form of Hyper IgE syndrome. J Allergy Clin Immunology 124: 1289-1302.

3. Freeman AF, Holland SM (2009) Clinical Manifestations, etiology and pathogenesis of the Hyper-IgE syndromes. Pediatric Res 65: 32-37.

4. Rael EL, Marshall RT, McClain JJ (2012) The Hyper-IgE Syndromes: Lessons in Nature, from Bench to Bedside. World Allergy Organ J 5:79-87.

5. Singh A, Mandal A, Seth R (2016) Hyper IgE Syndrome: often a missed diagnosis. Into J Contempt Pediatric 3: 674-677.

6. Grimbacher B, Holland SM, Puck JM (2005) Hyper-IgE syndromes. Immunology Rev 203: 244-250.

7. Wakim M, Alazard M, Yajima A, et al. (1998) High dose intravenous immunoglobulin in atopic dermatitis and hyper-IgE syndrome. Ann Allergy Asthma Immune 81: 153-158.

8. Ghaffari J, Ahanchian H, Zandieh F (2014) Update on Hyper IgE syndrome (HIES). J Pediatric Rev 2.

9. Yasharpour MR, Agarwal S, Jerome D (2014) Is it Hyper IgE Syndrome or Something Else? MOJ Immunology 1: 00010-00014.

10. Klion AD, Bochner BS, Gleich GJ, et al. (2006) Approaches to the treatment of Hypereosinophilia syndromes: a workshop summary report. J Allergy Clin Immunology 117: 1292.

**Citation:** Fehmida Najmuddin (2017) Hyper-IgE Syndrome: A Rare Entity. SF J AIDS HIV Res 1:1.