Original Research Paper

Volume - 14 | Issue - 04 | April - 2024 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Paediatrics

HYPER IgE SYNDROME – A CASE REPORT.

Dr. Shaik Irfani 3rd year Post Graduate, M.D. Paediatrics

Dr. P. Fathima BI

KEYWORDS :

INTRODUCTION

Hyper IgE Syndrome is a group of primary immunodeficiency disorders with overlapping and distinct immunologic and non - immunologic features . It has two types of presentations. Type 1 is an autosomal dominant disorder with STAT 3 mutation. Type 2 is an autosomal recessive type with DOCK 8 mutation.

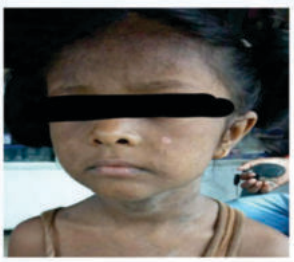
Case Report

A 6 year old female presented to us with % fever,cough and mild chest pain for 1 month.

Past history: There was history of similar episodes in the past with 4-5 hospital admissions where she was treated with antibiotics and previous hospital admission for 10 days 1 month back with pus accumulation in chest where an ICD was inserted .There was history of retained primary teeth.

Clinical Examination

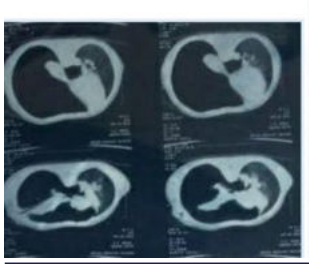
On examination :
Vitals: HR -98/min, RR -34/MIN ,Spo2-95% on RA.
Chest -decreased air entry of right side.
Rest of the systemic examination was normal .
L/E : multiple pustules on the neck and trunk.

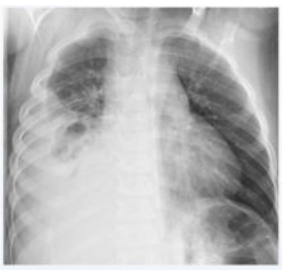


Investigations :

- Hb: 11.4,TLC -8400 (p 70, L 26),AEC=700.
- LFT : normal ,KFT : normal CRP> 24 mg/dl
- Blood culture : sterile
- X ray chest - Homogenous opacity on right side.
- Ultrasound chest _ loculated collection with internal echoes in right pleural cavity with a depth of 2 cm.
- CECT chest right sided multiple emphysematous bullae with loculated pleural effusionDue to strong suspicion for immuno deficiency (HIES/CGD) ,we worked up for the same .
- Serum IgE =7064 IU/ml ,rest Ig levels normal. HIV -NON REACTIVE
- STAT 3 MUTATION -POSITIVE.

The NIH clinical feature scoring system had AD -HIES scoring of 40 points. So the diagnosis of HIES was made.





Treatment:

IV antibiotics and ICD insertion.

Discharged on prophylactic antibiotics (FLUCONAZOLE and SEPTRAN) and follow up. The patient was referred for curative treatment /HSCT at a specialized center.

DISCUSSION

HYPER Ig E syndrome is a rare primary immunodeficiency disorder characterized by recurrent eczema, skin abscesses (cold abscesses) , lung infections , eosinophilia and increased serum IgE levels.
INCIDENCE : 1: 10 OOOOO

- Affects males and females equally, children can become symptomatic within the first few days to weeks of life .
- STAT 3 mutation is characterized by mucocutaneous candidiasis, typical facial appearance, hyperextensibility joints ,retained primary teeth ,bacterial pneumonia and recurrent infections.
- DOCK 8 mutation characterized by recurrent viral infections , allergic manifestations, malignancies and neurological complications.
- Diagnosis is based on the HIES scoring system developed at NIH in which patients are evaluated for the existence and severity of clinical and laboratory features
- Antimicrobials and IgG replacement are essential in the care for patients with STAT 3 MUTATION , whereas HSCT is curative in DOCK 8 mutation

CONCLUSION

Hyper IgE syndrome can present as empyema thoracis and must be considered a possibility in recurrent cold abscesses and empyema.

OUTCOME:

early diagnosis and treatment of infections give good outcomes in patients with AD-HIES . But early consideration of HSCT should be made in patients with AR-HIES due to its more severe nature.

REFERENCES

1) Freeman AF, Holland SM. Clinical manifestations of hyper IgE syndromes. *Dis Markers* 2010;29:123–30.
2) Yong PF, Freeman AF, Engelhardt KR, et al. An update on the hyper-IgE syndromes. *Arthritis Res Ther* 2012;14:228.
3) Szczawińska-Popłonyk A, Kycler Z, Pietrucha B, et al. The hyperimmunoglobulin E syndrome: clinical manifestation diversity in primary immune deficiency. *Orphanet J Rare Dis* 2011;6:76.
4) Farmand S, Sundin M. Hyper-IgE syndromes: recent advances in pathogenesis, diagnostics and clinical care. *Curr Opin Hematol* 2015;22:12–22
5) Grimbacher B, Holland SM, Gallin JI, et al. Hyper-IgE syndrome with recurrent infections: an autosomal dominant multisystem disorder. *N Engl J Med* 1999;340:692–702
6) Moin M, Farhoudi A, Movahedi M, et al. The clinical and laboratory survey of Iranian patients with hyper-IgE syndrome. *Scand J Infect Dis* 2006;38:898–903.