International Journal Dental and Medical Sciences Research
Volume 3, Issue 3, May - June 2021 pp 385-386 www.ijdmsrjournal.com ISSN: 2582-6018

CASE REPORT- HYPER IGE SYNDROME

1) Dr.Pritha Ghosh, 2) Dr.Raveesh Kumar
(Junior Resident, Department of Pediatrics, Burdwan Medical College and Hospital, West Bengal)
(Junior Resident, Department of Pediatrics, Burdwan Medical College and Hospital, West Bengal)

Submitted: 01-06-2021 Revised: 16-06-2021 Accepted: 18-06-2021

ABSTRACT: Hyper IgE syndrome is a rare, primary immunoregulatory disorder characterised by early-onset atopy and recurrent skin and lung infections.[1]
In this case report we present a case of Hyper IgE syndrome who underwent treatment along multiple lines before a diagnosis of Hyper IgE syndrome was reached.

I. INTRODUCTION

David et al. first described "Job Syndrome" in 1966 in two patients with eczema,recurrent pulmonary infections , and cold lung abscesses. Later , in 1972, Buckley et al. reported the association of this condition with increased serum immunoglobulin E (hyper IgE) levels.[2]

The autosomal dominant hyper IgE syndrome is caused by heterozygous mutations in the gene encoding signal transducer and activator of transcription 3(STAT-3).These mutations result in a dominant negative effect.[3]
There has been very few documented cases of Job's syndrome in the world and this case report intends to shed some light on why it is so and the maze of investigative workup done until the diagnosis was reached.

II. CASE REPORT

A 10 year old boy was admitted to the Paediatrics ward of Burdwan Medical College and hospital with the complaint of multiple oral ulcerations and bleeding through them. There was a history of recurrent abscesses over whole body since the age of 6 months, occurring every once or twice in a month , for which the patient used to consult local physician and subsided on antibiotics.

Prior history of admission in Hospital 7 months back for loculated pleural effusion for which the patient was started on antibiotics. Birth history and ante-natal history was uneventful. The child was fully immunised .There was no history of intellectual impairment,no history of developmental delay. The child does not attend school due to the troublesome features.

No other significant family history apart from the undiagnosed death of his father 10 months back, who had complaints of fever and hemoptysis, and unfortunately expired before his condition could be diagnosed.

On examination, the patient was alert , conscious and cooperative. GCS-15/15. General examination revealed multiple oral ulcerations in various stages of healing with rough , irregular surface of tongue and high arched palate. There was facial dysmorphism (deep set eyes, broad nasal bridge, rough and dry appearance of facial skin) .Skin over the whole body wasdry ,scaly with signs of healed abscesses.
On admission, detailed investigative workup was done with a suspicion of immunodeficiency:
Mantoux- Negative
CBNAAT for sputum- Negative
HIV-1 &2- Negative
HBsAg, anti-HCV- Negative
Complete Blood Count revealed-
increasedPlatelet count (7.54 lakhs/mm3) ,
elevatedEosinophil count(8%),and increased ESR(37)
Peripheral Blood Smear- did not find any abnormal cells
CRP-18, Serum Electrolytes- Normal
Kidney, Liver function tests- Normal
Prothrombin Time , INR, aPTT- was normal
Serum Vit D3 was marginally decreased
Serum IgE – 4075 ng/ml
Serum IgG – 20.4
Chest X ray and ECG – were normal

Thus the patient was diagnosed as Hyper IgE syndrome and necessary counselling was done to improve the quality of life.

III. DISCUSSION-

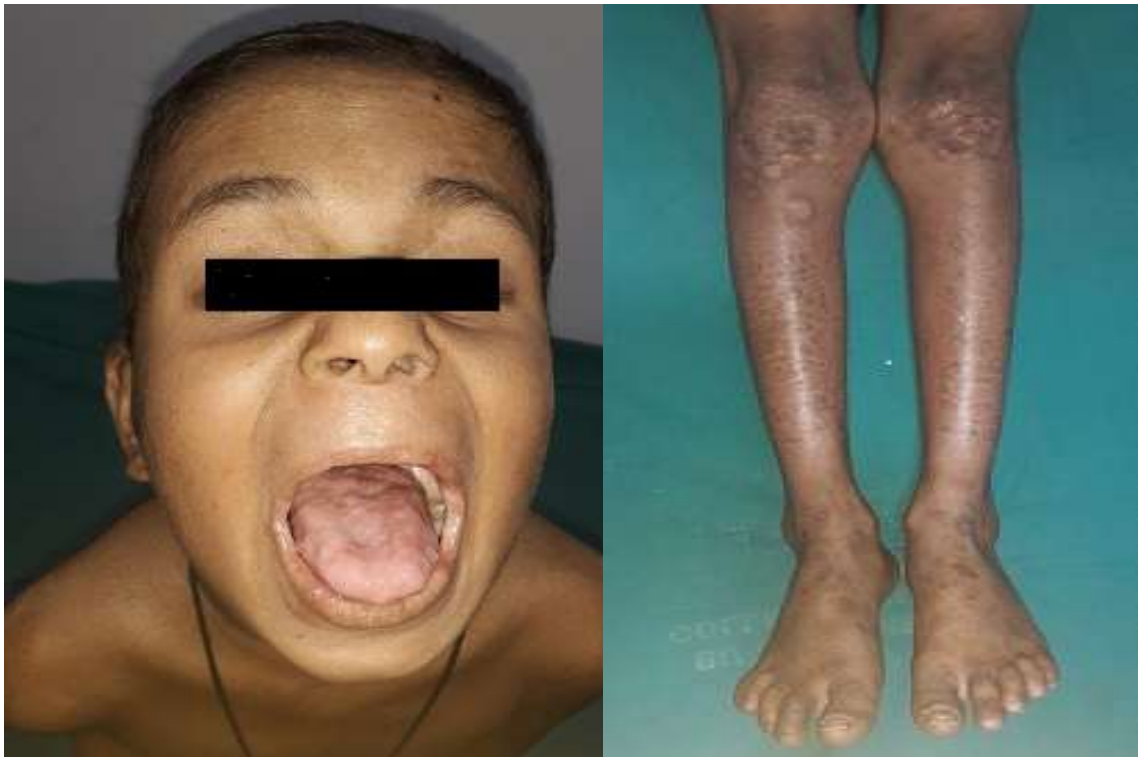
Hyper IgEsyndrome , also known as Job's syndrome and Buckley syndrome is characterised by skin abscesses, recurrent pneumatoceles or staphylococcal infections, high serum Ig E levels and eczematous dermatitis. It can be divided into two variants- Autosomal Dominant (due to STAT 3 gene) and Autosomal Recessive (due to DOCK8 gene mutation).[4]

Elevated serum levels of Ig E along with history of recurrent skin, oral and pulmonary

infections since infancy points to the diagnosis of Hyper IgE syndrome. Genetic analysis facility was not available in this region.

Therapeutic strategy is aimed at prevention and management of recurrent infections and counselling and aiding in resumption of day-to-day activities.

Other associated findings in this case- low serum Vit D3, increased platelet count, increased ESR, increased eosinophil count need further studies for determination of strength of association and any clinical correlation, if any.



REFERENCES-

[1]. Gupta P, Menon PS, Ramji S, Lodha R. PG Textbook of Pediatrics. 2nd ed. New Delhi:Jaypee;2018.193p.

[2]. Ameta P, Dhyani A, Nayak VH, Goyal S. Hyper IgE Syndrome: A case report. NitteUniv J of Health Science 2015 Dec;5(4):99-101.

[3]. Sullivan KE, Buckley RH. Autosomal Dominant Hyper-IgE Syndrome. In: Kliegman, Geme S, Blum, Shah, Tasker, Wilson (eds) Nelson Textbook of Pediatrics.21st ed. Philadelphia:Elsevier;2020.1118p.

[4]. Singh A, Khera K, Karanam A, Chauhan S. Job's syndrome in a pediatric patient: Case report and the clinical review. Indian J PaediatrDermatol 2015;16:96-8