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Case Report

Hyper-IgE syndrome with recurrent infections — An autosomal dominant multisystem disorder

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ABSTRACT

Hyper IgE syndrome (HIES) is a rare disorder characterized by eczema, recurrent infections of the skin and lungs, typically with Staphylococcus aureus, Candida albicans and certain viruses, and elevated levels of serum IgE. Other clinical manifestations include characteristic facies (prominent forehead, broad nasal bridge and facial asymmetry), chronic eczematous dermatitis, retained primary dentition, recurrent pathological fractures, hyper-extensibility and scoliosis. The respiratory system involvement in HIES has been reported with HRAD. Here we presented a case of HIES with rare associations of Hyperreactive airway disease.

1. Introduction

Hyper Ig E syndrome (HIES), first defined in 1966 by Davis,[1] is a multisystem disorder characterized by eczema, recurrent skin and pulmonary infections and markedly increased levels of serum Ig E. Other clinical manifestations included characteristic facies (prominent forehead, broad nasal bridge and facial asymmetry), retained primary dentition, hyperextensibility and scoliosis. The majority of cases occur sporadically; however, two types of HIES, the autosomal dominant (AD-HIES) form and the autosomal recessive form (AR-HIES), have also been reported. Reports of the central nervous system (CNS) involvement are rare in both types of HIES. Here we present a patient with HIES who developed Hyper reactive airway disorder.

1.1. Case Report

A 6 year old male child born of non-consanguineous marriage, only child, father is a k/c/o scoliosis was brought to emergency room with history of cold, cough since 2 days shortness of breath since the evening on the day of admission, child was admitted in picu initial investigations done and medically managed as HRAD, no similar complaints in the past. Past history of recurrent skin lesions since 1 year of age pustular lesions started on face spreaded to whole body associated with itching, pus discharge oozing pus c/s showed different bacterial growth, medically managed, healed by scarring. H/o inappropriate weight gain since 1 year of age. H/o eczema more during winters since 2 years of age. No h/o fever, evening rise of temperature, abdominal pain, vomiting, loss of appetite, altered bowel movements, weight loss, medication, trauma, allergy. On examination: Child is alert, appears to be malnourished Vitals: stable Head to toes Shape of the head normal Hair: thin hair pattern, omphysis pattern, atopic scarring, healed lesions Triangular facies multiple scars over face and body with fresh and healed lesions No buccal pad of fatAngular cheilosis, stomatitis Low set ears present Retention of primary dentition Systemic examination: NormalInvestigations: CBP:Hb:15.5Pcv: 45Tlc: 6800Plt: 2.55LNormal in morphology, maturity, distribution. Wbc:

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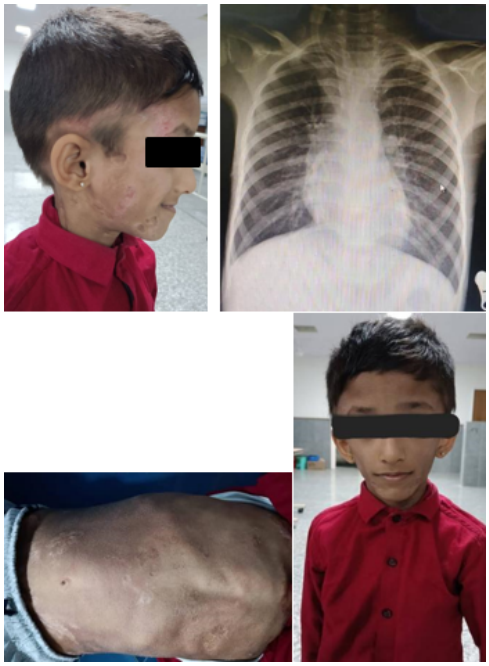

Fig. 1:

relative eosinophilia Platelet: adequate N 47, L 38, E 9, M 5, B 1CRP Negative ESR: 4mmImmunoglobulin Ig E (IgE) >3000PUS for culture sensitivity shows growth of pseudomonas auroginosa.

2. Discussion

HIES is a multi systemic disorder characterized by eczema, recurrent pulmonary and skin infections and markedly increased levels of serum IgE. The majority of cases occur sporadically; however, two types of HIES have also been reported. AD-HIES, caused by mutations in STAT3 identified in 2007,[2] presents with skeletal, connective tissue, and pulmonary abnormalities by whereas, AR-HIES, caused by mutations in DOCK8, identified in 2009,[3] manifests as severe eczema, recurrent bacterial and viral skin infections.[4]

Our patient had recurrent skin lesions over face, trunk, back which healed with cribriform scarring, child underwent skin grafting over trunk ulcer at age of 1 year, 1 year later child presented with non healing solitary scar over lower abdomen initially started as lesion that raised to ulcer since 7 days, biopsy showed suppurative inflammation in dermis with ulceration of overlying epidermis- pyoderma gangrenosum was made, Was medically managed with gentamycin and Dapson. Child has classical traingular facies.[5–7]

With healed lesions over body with atopic dermatitis, Retention of primary dentition. Child was regularly counseled regarding nutrition. At 5 years of age child was presented with hyperreactive airway disease was medically managed. The patient was not developed any epilepsy disorder.

Our patient has history of recurrent infections, we speculate that the cause is associated with elevated total serum IgE, which have functional attributes, both direct and indirect, serving to defense by limiting invasion of infective agents such as bacteria and viruses at the vulnerable mucosal surfaces.

The coexistence of growth retardation was present in our child, psychomotor delay, epilepsy and talipes cavus are rare and were not associated.

Currently child is well healthy going to school.

3. Conflicts of Interest

All contributing authors declare no conflicts of interest.

4. Source of Funding

None.

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