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Case Report

Recurrent pulmonary infections with pneumatocele and mucocutaneous involvement as presenting features of STAT3 hyper-IgE syndrome: a pediatric case report

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ABSTRACT

Hyper-IgE syndrome (HIES) is an inborn error of immunity characterized by recurrent infections, dermatitis, markedly elevated serum IgE and, in STAT3-associated disease, characteristic facial and dental abnormalities. We report a 5-year-5-month-old girl with recurrent otorrhea, oral candidiasis, respiratory infections and chronic skin lesions who presented with pneumonia and respiratory distress. Examination showed grade II clubbing, broad forehead, depressed nasal bridge, widely spaced teeth with dental caries, dermatitis and onychomycosis. Investigations demonstrated leukocytosis, eosinophilia, elevated inflammatory markers and serum IgE >3000 IU/mL, with normal lymphocyte subsets and a normal dihydrorhodamine assay. Chest imaging showed right upper-lobe cavitary disease followed by development of a large multiseptated pneumatocele measuring 93 × 99 × 68 mm. Whole exome sequencing confirmed STAT3-associated Hyper-IgE syndrome. The child improved with oxygen and intravenous ceftriaxone, with resolution of respiratory distress. This case highlights the importance of recognizing the characteristic mucocutaneous, dental and pulmonary features of STAT3-associated HIES in children with recurrent infections and emphasizes the role of molecular diagnosis.

Keywords: Hyper-IgE syndrome, STAT3, Inborn errors of immunity, Recurrent pneumonia, Pneumatocele, Onychomycosis

INTRODUCTION

Hyper-IgE syndrome (HIES) is a rare inborn error of immunity characterized by recurrent bacterial and fungal infections, dermatitis, markedly elevated serum IgE and characteristic non-immunologic manifestations. STAT3-associated HIES is associated with recurrent pulmonary infections and structural lung complications, including pneumatoceles, as well as characteristic facial, dental and skeletal abnormalities.[1-3] Recognition of the clinical phenotype is important because recurrent infections may initially be attributed to common childhood illnesses, delaying evaluation for an underlying immune disorder. Indian multicentre data demonstrate considerable clinical heterogeneity and support the value of molecular diagnosis in suspected HIES.[4] Pulmonary complications, particularly pneumatoceles, are an important cause of morbidity.[5] We describe a child with recurrent respiratory and mucocutaneous infections, chronic dermatitis, onychomycosis, clubbing, characteristic facial and dental findings, marked elevation of serum IgE and a large pulmonary pneumatocele, in whom whole exome sequencing confirmed STAT3-associated HIES.

CASE REPORT

A 5-year-5-month-old girl presented with cough for 1 month, fever for 10 days and fast breathing for 2 days. The cough was insidious in onset, gradually progressive and productive, with small quantities of mucoid, non-foul-smelling sputum. She had recurrent right ear discharge from 9 months of age, occurring every 4–5 months, and recurrent oral thrush from 1 year of age, occurring every 3-4 months. She had approximately 10–11 outpatient visits annually and recurrent admissions from 2 years of age for febrile respiratory or gastrointestinal illnesses. She had two admissions for pneumonia with acute gastroenteritis in January 2026 and further admissions for pneumonia in March and April 2026. Recurrent skin lesions had been present since 3 years of age.



Figure 1: Clinical features of the patient (A) Widely spaced teeth, (B) Dental caries, (C) Onychomycosis, (D) Well-defined erythematous scaly plaques over the back.

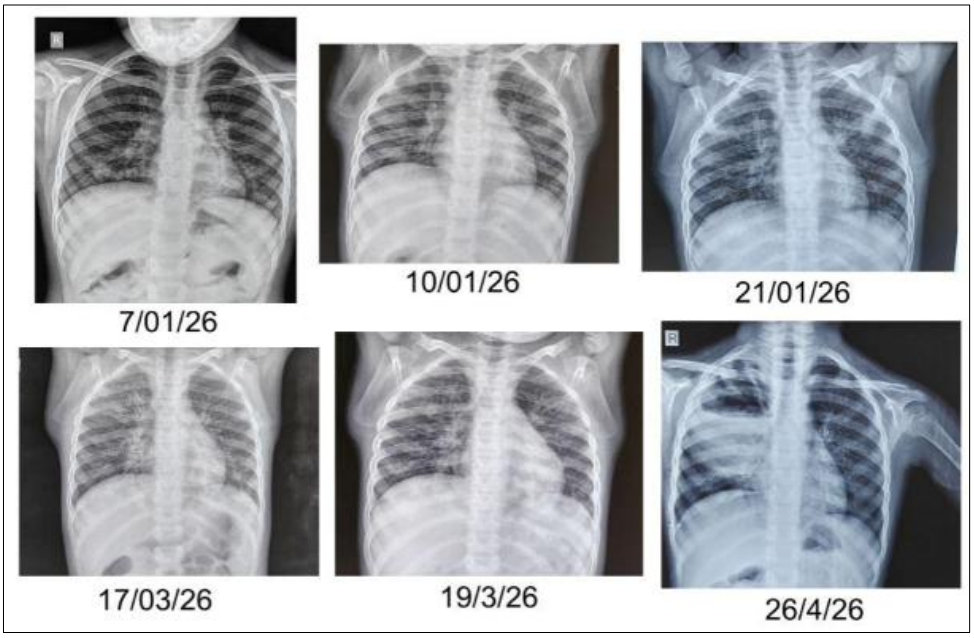


Figure 2: Serial chest imaging demonstrating right-sided pulmonary disease and development of a large right upper-lobe pneumatocele.

On admission, temperature was 98.4°F, pulse 96/min, respiratory rate 45/min, blood pressure 90/60 mmHg and oxygen saturation 88% on room air. Grade II clubbing was present in all four limbs. Facial examination showed a broad forehead, depressed nasal bridge and widely spaced teeth with dental caries. Yellow-green discoloration with subungual hyperkeratosis involved the left third finger and left great toe. Multiple erythematous to hyperpigmented papules and plaques with excoriation and crusting were present over the back. Height was 100 cm (−1.96 SD), weight 13 kg (−2.02 SD) and BMI 13 kg/m² (−0.95 SD).

Respiratory examination showed decreased vocal fremitus, dullness to percussion and decreased breath sounds over the right mammary and axillary areas. Cardiovascular, abdominal and neurological examinations were otherwise unremarkable.

Investigations showed hemoglobin 10.5–9.9 g/dL, leukocytosis of 17,000–18,480/mm³, thrombocytosis of 2.80–6.13 lakh/mm³, absolute eosinophil count 1360–1848/mm³, ESR 80 mm/h and CRP 38.7 mg/L. HIV, Mantoux test and sputum CBNAAT were negative. Sputum Gram stain showed gram-positive diplococci, while sputum and blood cultures showed no growth. Skin KOH examination was negative; nail KOH demonstrated hyaline, septate, branched filamentous hyphae. Lymphocyte subsets were normal, serum IgG, IgA and IgM were 1563, 104 and 168 mg/dL, respectively, and serum IgE was >3000 IU/ml. Dihydrorhodamine assay was normal.

Table 1: Complete blood count and inflammatory markers.

Parameters	01/05/2026	06/05/2026
Hemoglobin (g/dL)	10.5	9.9
WBC (/mm³)	17,000	18,480
Platelets (lakh/mm³)	2.80	6.13
ANC (/mm³)	11,390	9,240
ALC (/mm³)	4,080	6,098
AEC (/mm³)	1,360	1,848
ESR (mm/h)	80	—
CRP (mg/L)	38.7	—

Table 2: Immunological evaluation.

Investigations	Result	Reference/Interpretation
CD3+ cells	3,643 (74.7%)	Normal
CD4+ cells	2,170 (44.5%)	Normal
CD8+ cells	1,485 (30.4%)	Normal
CD4/CD8 ratio	1.4	Normal
CD19+ cells	920 (18.1%)	Normal
CD16/CD56+ cells	162 (4.6%)	Normal
IgG (mg/dL)	1,563	—
IgA (mg/dL)	104	—
IgM (mg/dL)	168	—
IgE (IU/ml)	>3,000	Markedly elevated
Dihydrorhodamine assay	Normal	No abnormality detected

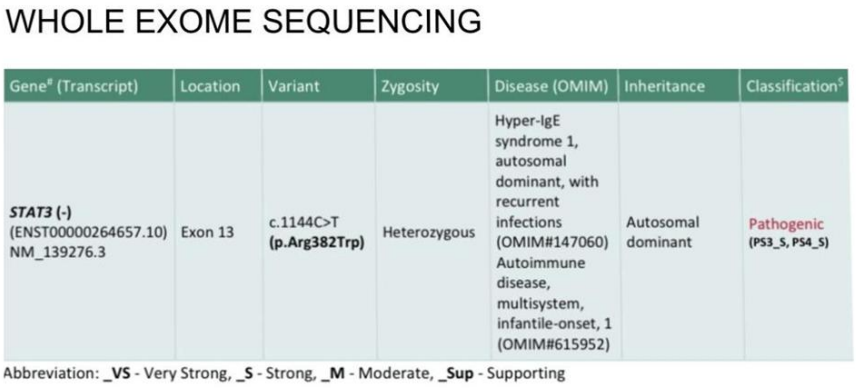


Figure 3: Whole-exome sequencing demonstrating the pathogenic STAT3 variant.

HRCT chest dated 02/05/2026 showed a cavitary lesion with air-fluid levels and right upper-lobe consolidation with bulging fissure, along with patchy right middle-lobe consolidation. The latest HRCT dated 11/06/2026 demonstrated a large air-filled multiseptated right upper-lobe cyst measuring 93×99×68 mm, with adjacent collapse-consolidation and patchy middle-lobe ground-glass opacities, consistent with a large pneumatocele. Whole exome sequencing confirmed STAT3-associated Hyper-IgE syndrome.

The child was treated with oxygen by face mask, symptomatic treatment and intravenous ceftriaxone. She became afebrile from the second day; antibiotics were continued for 7 days. Respiratory distress resolved and oxygen was discontinued after 7 days, with respiratory rate 26/min and oxygen saturation 96% on room air.

DISCUSSION

STAT3-associated HIES is a rare inborn error of immunity caused by heterozygous pathogenic variants in STAT3 and is characterized by recurrent bacterial infections, eczema or dermatitis, mucocutaneous candidiasis, markedly elevated IgE and characteristic skeletal, dental and facial abnormalities.[1-4] The present child had a characteristic combination of recurrent respiratory and mucocutaneous infections beginning early in childhood, chronic dermatitis, onychomycosis, eosinophilia, marked elevation of IgE and characteristic facial and dental findings. The normal quantitative lymphocyte subsets and normal dihydrorhodamine assay did not exclude HIES but helped narrow the differential diagnosis. Molecular confirmation by whole exome sequencing established the diagnosis.

Pulmonary disease is a major manifestation of STAT3-associated HIES. Recurrent pneumonia may lead to structural lung damage, including bronchiectasis and pneumatoceles.[3,5] In this child, serial imaging demonstrated cavitary right upper-lobe disease followed by formation of a large pneumatocele, while grade II clubbing suggested chronic pulmonary involvement. The combination of recurrent pneumonia, pneumatocele formation and characteristic extrapulmonary findings should prompt evaluation for HIES.

The differential diagnosis includes other inborn errors of immunity, particularly DOCK8 deficiency, chronic granulomatous disease and chronic mucocutaneous candidiasis, as well as tuberculosis and cystic fibrosis in appropriate clinical settings. The phenotype, markedly elevated IgE, normal dihydrorhodamine assay and characteristic facial and dental findings supported STAT3-associated HIES, which was subsequently confirmed genetically.

Management requires prompt treatment of infections, attention to skin and mucocutaneous disease and surveillance for pulmonary, dental and skeletal complications.[1,2] Early recognition is important to reduce recurrent infections and identify established pulmonary complications.

CONCLUSION

STAT3-associated Hyper-IgE syndrome should be considered in children with recurrent respiratory and mucocutaneous infections, markedly elevated IgE and characteristic facial or dental abnormalities. Pneumatocele formation and clubbing may indicate significant chronic pulmonary involvement. Recognition of the phenotype and molecular confirmation can facilitate appropriate long-term management and surveillance.

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