Case Report

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Paediatrics Section

Autosomal Dominant Hyper-IgE (Job) Syndrome Presenting with Recurrent Pulmonary Infections and Empyema in a Child: A Case Report

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ABSTRACT

Hyper-IgE syndrome (HIES), also known as Job syndrome, is a rare primary immunodeficiency characterised by recurrent cutaneous and pulmonary infections, chronic eczematous dermatitis, and markedly elevated serum immunoglobulin E levels. The Autosomal Dominant (AD) form, resulting from mutations in the Signal Transducer and Activator of Transcription 3 (STAT3) gene, is the most frequently encountered variant and is associated with significant infectious morbidity, particularly involving the respiratory system. The present case report describes a three-year-old male child who presented with a 10-day history of cough, tachypnoea, and right upper abdominal pain. He had a history of recurrent bilateral pneumonia, eczema, oral candidiasis, and abscess formation. Radiological evaluation revealed a right sided pleural empyema with substantial loss of lung volume. Laboratory investigations showed a markedly elevated serum IgE level of 2872 IU/mL, and genetic testing confirmed a STAT3 mutation, establishing the diagnosis of AD-HIES. Despite initial conservative management, the patient developed organising empyema and subsequently underwent Video-Assisted Thoracoscopic Surgical (VATS) decortication. The postoperative course was uneventful, with significant clinical and radiological improvement. Long-term antimicrobial prophylaxis was initiated. Follow-up was advised; however, the patient did not return for follow-up due to relocation. The present case underscores the importance of considering HIES in children with recurrent pulmonary infections and severe pleural complications, and highlights the role of early diagnosis, genetic confirmation, and timely surgical intervention in improving clinical outcomes.

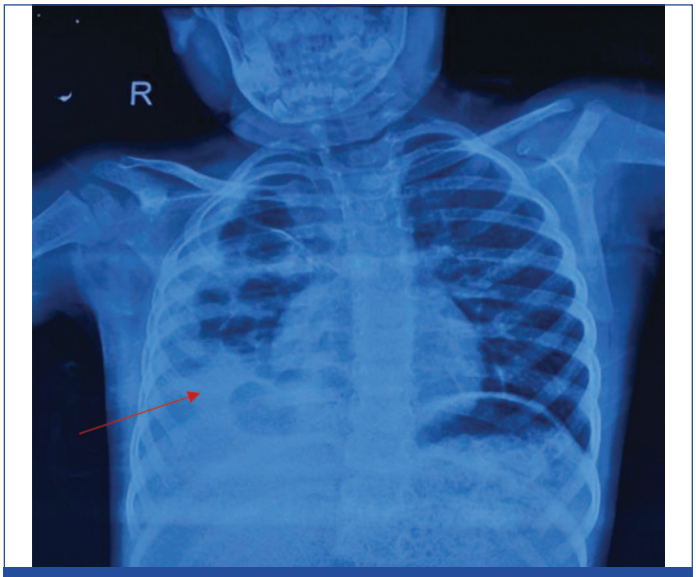
Keywords: Immunoglobulin E, Primary immunodeficiency, Signal transduction, Staphylococcal infections, Thoracoscopy

CASE REPORT

A three-year-old male child was referred to the Department of Paediatrics at a peripheral military hospital with complaints of fever, cough, fast breathing, and right upper abdominal pain for 10 days. The child was born preterm at 34 weeks of gestation by lower-segment caesarean section due to premature rupture of membranes, with a birth weight of 1.6 kg. The child required neonatal intensive care and was intubated for 10 days. On the first day of life, the child developed a right sided pneumothorax, for which an intercostal drain was inserted, and received surfactant therapy. The child was born to non consanguineous parents. There was no significant family history of similar illness or recurrent infections.

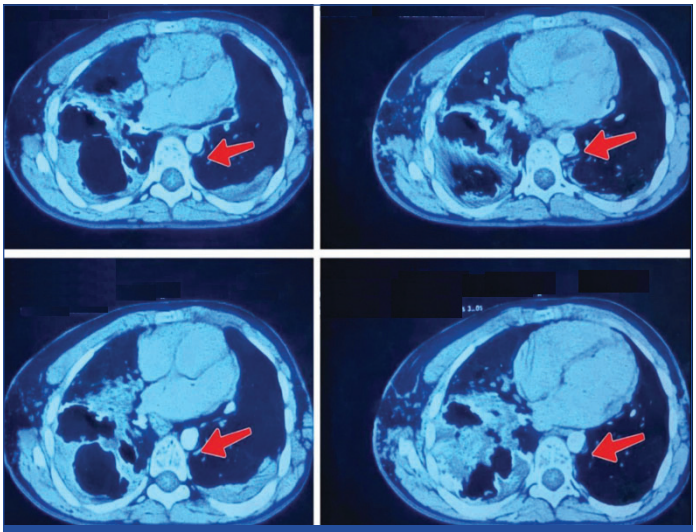
At seven months of age, the child developed difficulty in breathing associated with stridor and was diagnosed with bilateral pneumonitis, which was managed conservatively. At eight months, the patient had a similar episode requiring hospital admission for 15 days, which was again managed conservatively. At nine months, he presented with recurrent respiratory distress. Computed tomography of the thorax revealed focal severe subglottic stenosis, multifocal consolidation of the right lung, and mosaic attenuation in the left lung. Laryngoscopic evaluation confirmed Type II laryngomalacia, for which he underwent supraglottoplasty. He also had a history of an intra-abdominal abscess in early infancy, which required drainage at a military service hospital, with culture growing *Staphylococcus aureus*. In addition, the child had chronic eczema, oral thrush, and a chalazion involving the left upper eyelid.

On examination, the child was alert but irritable. Vital parameters revealed a heart rate of 142/min, respiratory rate of 58/min, blood pressure of 110/64 mmHg, oxygen saturation of 95% on room air, and temperature of 100°F. On general examination, pallor was present, with no cyanosis, clubbing, or icterus. A pigtail catheter was noted in situ on the right side of the chest. Skin examination revealed multiple exfoliating healing lesions. On respiratory examination, air entry was reduced on the right side. Clinical and radiological evaluation at presentation revealed a right sided pleural effusion [Table/Fig-1]. The patient was managed initially with pigtail catheter drainage and intravenous antibiotics, including meropenem (280 mg three times daily). Oral antimicrobial prophylaxis included cotrimoxazole (40 mg on alternate days), fluconazole (50 mg once daily), and penicillin G (2 lakh units twice daily).



[Table/Fig-1]: Chest radiograph at presentation. Initial chest X-ray showing right sided pleural opacity suggestive of pleural effusion with underlying pulmonary involvement (arrow indicating homogeneous opacity in the right lower zone).

A Non Contrast Computed Tomography (NCCT) scan of the chest demonstrated empyema involving the entire right hemithorax, with significant volume loss of the right lower lobe [Table/Fig-2]. In view of the recurrent history of atopic dermatitis, recurrent skin and pulmonary infections, and abscess formation, an underlying primary immunodeficiency was suspected. Laboratory investigations revealed markedly elevated serum immunoglobulin E levels of 2872 IU/mL. Genetic evaluation confirmed the presence of a STAT3 mutation, establishing the diagnosis of AD-HIES.



[Table/Fig-2]: NCCT chest showing empyema. NCCT scan of the chest demonstrating empyema involving the entire right hemithorax with significant volume loss of the right lower lobe (red arrow).

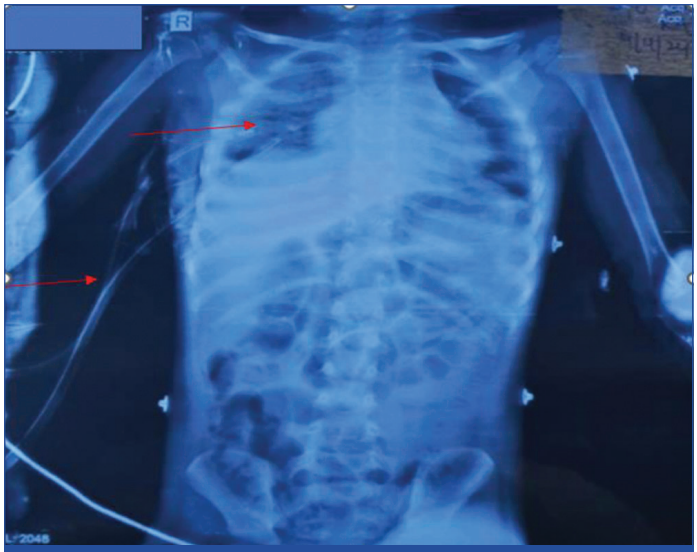
Following initial medical stabilisation and optimisation, the patient underwent preanaesthetic evaluation, which revealed stable haemodynamic status with adequate respiratory function and no contraindications to surgery. The patient underwent VATS decortication in the left lateral decubitus position with the right side up. A camera port was introduced through the previous drainage site, followed by placement of two additional working ports under direct vision. Thoracoscopic evaluation revealed an empyema cavity with thick purulent material predominantly in the basolateral region of the right lung, along with necrotic tissue, which was debrided. Fibrinous peel was removed, facilitating lung reexpansion. Two intercostal chest drains were placed on the right side. Postoperatively, the patient was managed with intravenous antibiotics, including vancomycin (210 mg four times daily) and meropenem (280 mg three times daily) for two weeks, along with antimicrobial prophylaxis using cotrimoxazole (40 mg on alternate days), fluconazole (50 mg once daily), and penicillin G (2 lakh units twice daily). On discharge, the patient was continued on oral cotrimoxazole (40 mg on alternate days), fluconazole (50 mg once daily), and penicillin G (2 lakh units twice daily).

Postoperative chest radiography showed improved lung expansion with intercostal drainage tubes in situ following video-assisted thoracoscopic decortication [Table/Fig-3].

The patient demonstrated good clinical recovery during the postoperative period. Incentive spirometry was encouraged, with satisfactory improvement in respiratory function. The patient was discharged after 13 days of hospitalisation on oral antimicrobial prophylaxis including cotrimoxazole (40 mg on alternate days), penicillin G (2 lakh units twice daily), and fluconazole (50 mg once daily) for 30 days. Follow-up was advised after one week; however, the patient did not return for follow-up as he was relocated to another state.

DISCUSSION

The AD-HIES, also known as Job syndrome, is a rare primary immunodeficiency characterised by recurrent infections and



[Table/Fig-3]: Postoperative chest radiograph. Chest X-ray following Video-Assisted Thoracoscopic Surgical (VATS) decortication showing improved lung expansion and resolution of pleural pathology (red arrow).

multisystem involvement resulting from underlying immune dysregulation. First described by Davis SD et al., in 1966 and later characterised by Buckley RH et al., with the identification of elevated serum IgE levels, the syndrome remains a diagnostic challenge due to its rarity and heterogeneous clinical presentation [1-3]. Comprehensive reviews and clinical descriptions have emphasised the wide phenotypic variability observed in affected patients, particularly in paediatric populations [4].

The AD-HIES is most commonly caused by mutations in the STAT3 gene, which encodes the signal transducer and activator of transcription-3, a key regulator of immune signaling pathways [5-8]. STAT3 mutations impair T-helper 17 (Th17) cell differentiation, leading to defective neutrophil recruitment and reduced inflammatory responses at sites of infection. This immunological defect predisposes patients to recurrent bacterial and fungal infections, particularly involving the skin and respiratory tract [1,5]. In the present case, genetic testing confirmed a STAT3 mutation, providing a definitive diagnosis and explaining the early onset and recurrent nature of infections observed since infancy.

Clinically, AD-HIES is classically characterised by markedly elevated serum IgE levels, eczematous dermatitis, and recurrent staphylococcal infections of the skin and lungs [1,3]. The present patient exhibited all hallmark features, including early-onset eczema, recurrent pneumonia, recurrent abscess formation with *Staphylococcus aureus* isolation, and a significantly elevated serum IgE level exceeding 2000 IU/mL. A paediatric case reported by Nagaraj P et al., described similar recurrent infections and delayed diagnosis of HIES, highlighting that such cases are often misdiagnosed as severe atopic disease, leading to delayed referral and progression to complications [6].

Pulmonary involvement is a major determinant of morbidity and long-term outcome in AD-HIES. Recurrent pneumonias caused predominantly by *Staphylococcus aureus* and Haemophilus influenzae may progress to lung abscesses, pneumatoceles, bronchiectasis, and pleural complications [7]. Indian case series and reviews have documented a high burden of pulmonary sequelae in patients with HIES, underscoring the need for early intervention and close respiratory follow-up [7]. In the present case, repeated pulmonary infections culminated in organising empyema involving the entire right hemithorax, with significant loss of lung volume.

Empyema is known to progress through exudative, fibrinopurulent, and organising stages. While early stages may respond to antibiotic therapy and drainage, advanced organising empyema often requires surgical intervention to achieve lung reexpansion [8]. In the present case, failure of conservative management necessitated VATS

decortication. Recent paediatric surgical literature supports early VATS as an effective and minimally invasive approach for managing advanced empyema, allowing complete evacuation of purulent material, removal of fibrous peel, and improved postoperative recovery when compared to open thoracotomy [9].

A large retrospective study involving 167 children undergoing VATS decortication demonstrated effective clearance of infection with good lung reexpansion, minimal postoperative complications, and shorter hospital stay, supporting its safety and efficacy in advanced empyema [10]. Similarly, a paediatric observational study evaluating children with empyema who underwent VATS showed favourable outcomes, including reduced morbidity, early chest tube removal, and shorter duration of hospitalisation, highlighting its advantage over conventional approaches in selected cases [11].

Long-term management of AD-HIES focuses on prevention of recurrent infections and minimisation of organ damage. Prophylactic antimicrobial therapy targeting *Staphylococcus aureus* remains a cornerstone of care, with antifungal prophylaxis considered in patients with recurrent candidiasis or severe pulmonary involvement [1,8]. In the present case, initiation of long-term antibacterial and antifungal prophylaxis, along with respiratory rehabilitation, resulted in favourable clinical recovery. Multidisciplinary follow-up and genetic counseling are essential, particularly given the AD inheritance pattern and the potential for progressive complications [12].

CONCLUSION(S)

The AD-HIES is a rare immunodeficiency condition associated with recurrent infections and severe pulmonary complications. It should be suspected in children with eczema, recurrent pneumonia, and abscess formation. Early diagnosis with genetic confirmation and timely intervention are essential to prevent disease progression.

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