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HERMANSKY–PUDLAK SYNDROME: A RARE CAUSE OF PROGRESSIVE INTERSTITIAL LUNG DISEASE IN A YOUNG ADULT

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ABSTRACT

Hermansky–Pudlak syndrome (HPS) is a rare autosomal recessive disorder characterised by oculocutaneous albinism, bleeding diathesis, and systemic complications such as pulmonary fibrosis. We report a 33-year-old man with oculocutaneous albinism who presented with a three-month history of progressive exertional dyspnoea and dry cough. Clinical examination revealed horizontal nystagmus, cutaneous albinism, and actinic keratosis. Pulmonary evaluation demonstrated a restrictive pattern on spirometry, and high-resolution computed tomography revealed features suggestive of interstitial lung disease. Genetic testing supported the diagnosis of HPS. The patient was managed with long-term oxygen therapy and considered for antifibrotic therapy, alongside counselling for lung transplantation. This case highlights the need for early recognition of HPS-associated pulmonary fibrosis and timely referral for advanced respiratory interventions.

KEYWORDS: *Hermansky–Pudlak syndrome; oculocutaneous albinism; pulmonary fibrosis; interstitial lung disease; long-term oxygen therapy; lung transplantation.*

ABBREVIATIONS

- HPS – Hermansky–Pudlak Syndrome
- ILD – Interstitial Lung Disease
- HRCT – High-Resolution Computed Tomography
- LTOT – Long-Term Oxygen Therapy

INTRODUCTION

Hermansky–Pudlak syndrome (HPS) is a genetically heterogeneous autosomal recessive disorder characterised by tyrosinase-positive oculocutaneous albinism, a bleeding diathesis related to the absence of platelet dense bodies, and various systemic complications including pulmonary fibrosis and granulomatous colitis.[1,2]

The underlying mutations affect proteins involved in the biogenesis of lysosome-related organelles, leading to progressive accumulation of ceroid lipofuscin in tissues.[3]

To date, more than eleven genetic subtypes of HPS have been identified, among which HPS-1, HPS-2, and HPS-4 are most strongly associated with progressive interstitial lung disease (ILD).[4] HPS-related pulmonary fibrosis typically manifests during the third or fourth decade of life and is the leading cause of mortality in affected individuals.[1,5] We report a young man with lifelong oculocutaneous albinism in whom progressive exertional dyspnoea led to the recognition of HPS-associated pulmonary fibrosis, illustrating the diagnostic pathway from clinical suspicion to genetic confirmation.

CASE REPORT

History

A 33-year-old man from Tenkasi, born of a consanguineous marriage, presented with a three-month history of progressively worsening breathlessness and dry cough. He had lifelong oculocutaneous albinism, a history of easy bruising, and a positive family history of albinism.

Clinical Examination

- Cutaneous albinism (Fig. 1)
- Horizontal nystagmus
- Iris transillumination
- Actinic keratosis (Fig. 2)
- Albinotic fundus (Fig. 3)

Respiratory examination revealed bibasilar fine crepitations.

INVESTIGATIONS

Investigation	Finding
Pulmonary function test	Restrictive ventilatory pattern
HRCT chest	Features suggestive of interstitial lung disease, consistent with HPS-associated pulmonary fibrosis (Fig. 4)
Genetic testing	Confirmed a pathogenic variant associated with Hermansky–Pudlak syndrome

MANAGEMENT AND OUTCOME

- Initiated long-term oxygen therapy (LTOT) for hypoxaemia.
- Antifibrotic therapy was considered, supported by recent reports of rapidly progressive HPS-related fibrosis.[6,7]
- The patient was counselled regarding lung transplantation, the only definitive therapeutic option for advanced HPS-related ILD.[1]
- Genetic counselling was advised regarding future pregnancies.

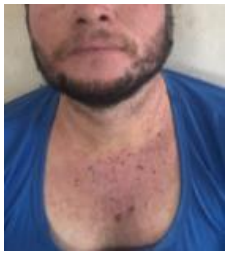

Fig. 1

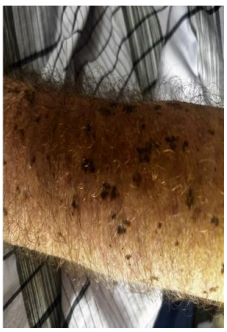

Fig. 2

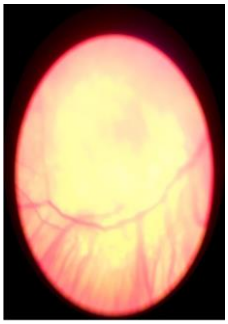

Fig. 3

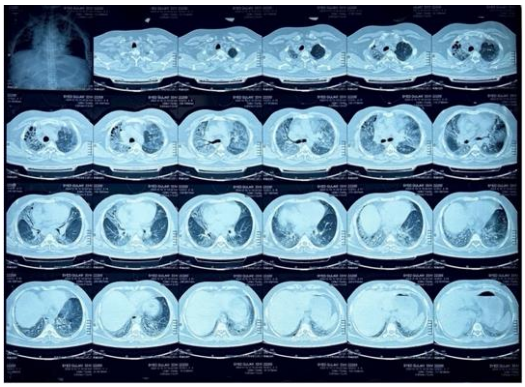

Fig. 4

Fig. 1: Cutaneous albinism. Fig. 2: Actinic keratosis. Fig. 3: Albinotic fundus. Fig. 4: HRCT chest showing features of interstitial lung disease.

DISCUSSION

HPS is a rare but clinically important cause of early-onset interstitial lung disease, with pulmonary fibrosis constituting the major cause of mortality. The pathogenesis involves abnormal lysosome-related organelle formation, leading to lipofuscin accumulation and progressive lung injury.[3,4]

Several case reports have shown that HPS-related pulmonary fibrosis may progress rapidly, sometimes presenting with complications such as pneumothorax and fulminant respiratory failure.[6,7] Early identification of ILD through spirometry, HRCT, and genetic confirmation is therefore essential.

Management remains largely supportive, with LTOT and antifibrotic agents offering symptomatic and modest disease-modifying benefit, respectively. Lung transplantation remains the only proven long-term survival-extending treatment in advanced cases.[1,5]

CONCLUSION

This case underscores the importance of recognising HPS in individuals with albinism who present with

respiratory symptoms, and highlights the need for timely referral for advanced lung disease management.

DECLARATIONS

Conflict of Interest: The authors declare no conflict of interest.
Patient Consent: Obtained.
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