Case Report

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An Unusual Presentation of Idiopathic Pulmonary Arterial Hypertension in a Young Male: A Case Report

Internal Medicine Section

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ABSTRACT

Idiopathic Pulmonary Arterial Hypertension (IPAH) is a progressive disease of the pulmonary vasculature without any known underlying risk factors. For the diagnosis Pulmonary Artery Hypertension (PAH), the mean Pulmonary Artery Pressure (mPAP) should be persistently more than 20 mmHg at rest. Early diagnosis, along with investigations like 2D Echocardiography (ECHO) and right heart catheterisation, and prompt treatment, are essential for the management of PAH. Drugs such as endothelin receptor blockers, phosphodiesterase inhibitors, and calcium channel blockers are used for treatment. Hereby, the authors reported an unusual case of IPAH in a young male, with haemoptysis being the predominant presenting complaint. A 24-year-old male patient presented with the primary complaints of streaky haemoptysis and dyspnoea. He had multiple hospital admissions for similar episodes of haemoptysis in the past, which had been managed symptomatically. After conducting a thorough investigation and detailed work-up, he was diagnosed with idiopathic PAH. The patient showed symptomatic improvement, with resolution of haemoptysis and dyspnoea, after initiating treatment.

Keywords: Haemoptysis, Pulmonary arteries, Right heart failure, Transoesophageal echocardiography, Young adult

CASE REPORT

A 24-year-old male shopkeeper presented to the Emergency Department with a two-week history of blood-tinged sputum and breathlessness, classified as grade 3 on the Modified Medical Research Council (mMRC) scale [1].

Upon reviewing his detailed history, the patient disclosed that he had been experiencing intermittent episodes of streaky haemoptysis for the past five years, leading to numerous hospital admissions. Each time, he was managed conservatively for haemoptysis with haemostatic agents such as ethamsylate and then discharged. The patient reported no symptoms of fever, chest pain, joint pain, abdominal pain, palpitations, syncope, or limb swelling. There was no history of substance abuse, and there was no significant family history.

The patient was conscious, oriented, and haemodynamically stable. During auscultation, bilateral suprascapular and infrascapular crackles were present. Examination of the cardiovascular system suggested loud P2 and an Ejection Systolic Murmur (ESM) in the pulmonary area.

Haemogram and biochemical investigations, such as renal function tests, liver function tests, and serum electrolytes, were normal [Table/Fig-1]. Other laboratory parameters, including Prothrombin Time (PT/INR), N-terminal pro b-type natriuretic peptide (NT-pro BNP), and thyroid profile, were within normal limits. The Human Immunodeficiency Virus (HIV) test was non reactive. The Rheumatoid Arthritis factor (RA factor) and Anti-nuclear Antibodies (ANA) profile were negative [Table/Fig-1].

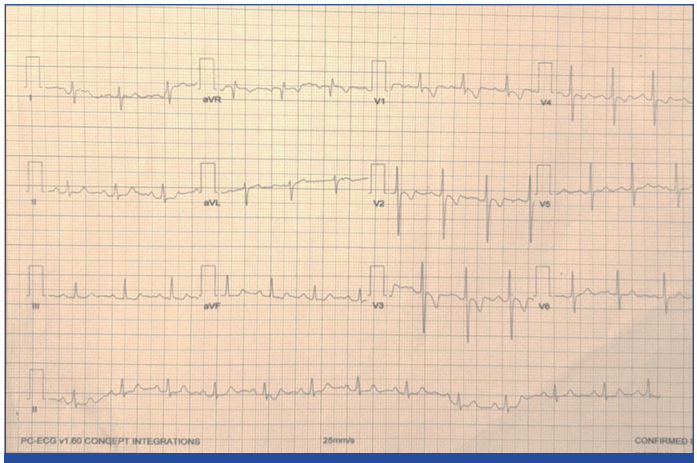
The ECG suggested T wave inversion in leads V1-V4 [Table/Fig-2].

Arterial Blood Gas Analysis (ABGA) indicated mild hypoxemia with a Partial Pressure of Oxygen (pO_2) of 75.7 mmHg. Sputum analysis, including cytology, Gram stain, cultures, and Cartridge-based Nucleic Acid Amplification Test (CBNAAT), returned normal results. The chest radiograph revealed an enlarged pulmonary hilum with normal parenchyma [Table/Fig-3].

A Transoesophageal Echocardiogram (TEE) was performed, which suggested severe PAH with a Right Ventricular Systolic Pressure (RVSP) of 116 mmHg, accompanied by dilation of the Right Atrium (RA), Right Ventricle (RV), and Main Pulmonary Artery (MPA). The

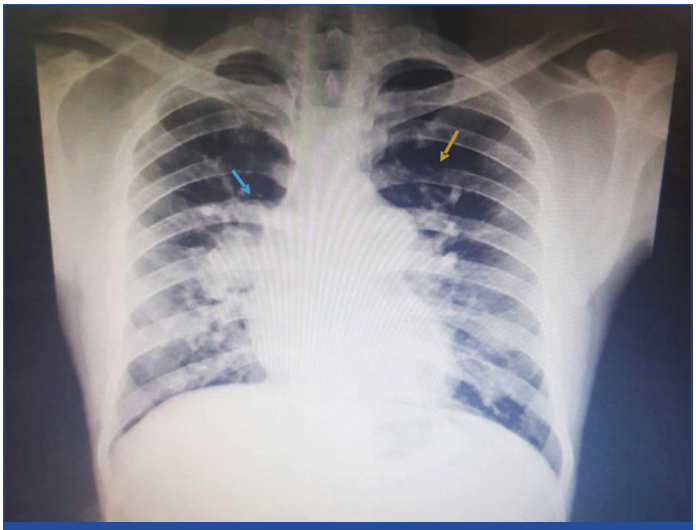
Investigations	Result	Normal range
Haemoglobin	15.7 g/dL	(13.5–17.5 g/dL for males)
Total leucocyte count	6,600/mm^3	(4,000-11,000/mm^3)
Platelets	2.4 Lacs/µL	(1.5–4.5 Lacs/µL)
Renal and liver function test	Normal	(Varies, all within normal range)
HIV	Non reactive	(Non reactive)
NT-proBNP	Normal	(<125 pg/mL for age <75 years)
Thyroid function test	Normal	(TSH: 0.4-4.2 µIU/mL)
RA factor/ANA profile	Negative	(Negative)
PT/INR	13.2 seconds/1.2	(PT: 11-13.5 sec; INR: 0.8-1.2)
Sputum studies	Normal	

[Table/Fig-1]: Laboratory investigations.
RA: Rheumatoid factor; ANA: Antinuclear antibody test; PT/INR: Prothrombin time/International normalised ratio; HIV: Human immunodeficiency virus; NT-proBNP: N-terminal pro-B-type natriuretic peptide

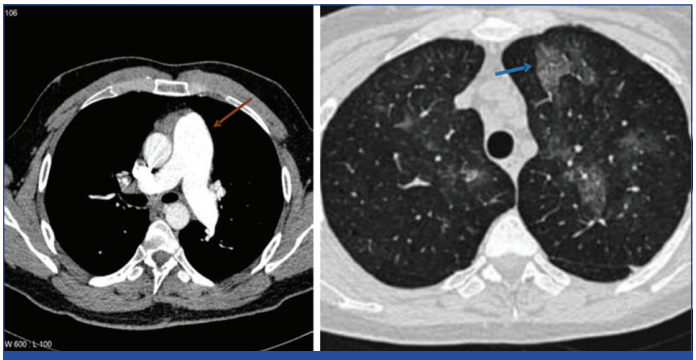


[Table/Fig-2]: ECG showing T wave inversion from V1- V4.

Left Ventricular Ejection Fraction (LVEF) was measured at 55%. A Computed Tomography Pulmonary Angiogram (CTPA) indicated a dilated MPA (38.5 mm) with patchy Ground-glass Opacity (GGO) in the bilateral upper lobes and no evidence of pulmonary thromboembolism [Table/Fig-4a,b].



[Table/Fig-3]: The yellow arrow shows a prominent pulmonary conus, the blue arrow shows enlarged pulmonary hila.



[Table/Fig-4]: CECT Thorax showing dilated main pulmonary arterial trunk (red arrow), Ground Glass Opacity (GGO) (blue arrow).

Polysomnography results were within normal limits, showing an Apnoea-Hypopnoea Index (AHI) of 1.3.

Spirometry was normal, demonstrating no obstruction or restriction. The diffusing capacity of the lung for carbon dioxide (DLCO) was also normal at 98%. A six-minute Walk Test (6MWT) revealed no significant desaturation.

Summary of the radiological investigations, along with pulmonary and cardiac evaluations has been depicted in [Table/Fig-5]. Every other possible cause of pulmonary hypertension, such as cardiac disease, pulmonary disease, deep vein thrombosis, HIV, connective tissue disorders, haematological disorders, pulmonary thromboembolism, and obstructive sleep apnoea, was ruled out, leading to the diagnosis of idiopathic PAH classified as WHO functional class 3 [1].

Chest radiograph	Prominent pulmonary conus with enlarged pulmonary hila with normal parenchyma
ECG	T wave inversion in V1-V4
2D ECHO	Severe PAH (RVSP 116), with dilated RA/RV and LVEF of 55%
ABGA	Mild hypoxemia (ph-7.41, pO_2-75.7, pCO_2-34.8, HCO_3-21.8)
6MWT	No significant desaturation (distance walked-420 m, pre SpO_2-95%, post SpO_2-98%)
DLCO	Normal (98%)
Polysomnography	WNL (AHI-1.3, no evidence of OSA)
Spirometry	Normal, No obstruction, No restriction (FEV1: 2.27 L, FEV1/FVC: 80.7%, FVC: 2.81 L)
USG abdomen/pelvis	No anomaly detected
Bilateral lower limb colur doppler	Normal
CTPA	No evidence of pulmonary thromboembolism

[Table/Fig-5]: Radiological investigations as well as pulmonary and cardiac evaluations.
ECG: Electrocardiogram; 2D ECHO: Two-dimensional echocardiography; PAH: Pulmonary arterial hypertension; RVSP: Right ventricular systolic pressure; RA/RV: Right atrium/right ventricle; LVEF: Left ventricular ejection fraction; ABGA: Arterial blood gas analysis; 6MWT: Six-minute walk test; SpO_2: Peripheral oxygen saturation; DLCO: Diffusing capacity of the lung for carbon monoxide; WNL: Within normal limits; AHI: Apnea-hypopnoea index; OSA: Obstructive sleep apnea; FEV_1: Forced expiratory volume in 1 second; FVC: Forced vital capacity; USG: Ultrasonography; CTPA: Computed tomography pulmonary angiography

The patient was commenced on oxygen therapy, alongside tablet Ambrisentan (endothelin receptor blocker) and tablet Tadalafil (phosphodiesterase-5 inhibitor) (5+20 mg) once daily; tablet Furosemide (diuretic) and tablet Spironolactone (potassium-sparing diuretic) (50 mg) twice daily; and tablet Digoxin (cardiac glycoside) 0.25 mg once daily, five days a week [2].

Additionally, the patient received vaccinations for pneumococcal and influenza infections. He was discharged after 10 days, having experienced symptomatic improvement with resolution of haemoptysis and breathlessness. The patient is undergoing routine follow-up every six months, and the most recent TEE indicated a decrease in RVSP to 51 mmHg (compared to the RVSP of 116 mmHg at the time of admission).

DISCUSSION

Pulmonary hypertension is defined as a mPAP of more than 20 mm Hg at rest [2]. PAH includes those patients who, in addition to having a mPAP of more than 20 mmHg, also have precapillary hypertension, characterised by a pulmonary vascular resistance of more than three Wood units and an end-expiratory pulmonary artery wedge pressure of less than 15 mmHg [3]. PAH can be categorised into three types: idiopathic, heritable, and PAH associated with conditions such as connective tissue disease and congenital heart disease, among others. IPAH refers to those sporadic cases without a positive family history, hereditary forms, or any associated conditions like connective tissue disease or congenital heart disease [3].

The IPAH is a rare and progressive disorder that is difficult to cure and can ultimately lead to right heart failure and premature death. It is a diagnosis of exclusion and accounts for more than 50% of all PAH cases [4].

The incidence of IPAH is very low, with approximately 4 to 6 cases per million people worldwide. In the United States, there are nearly 140 deaths per year attributed to IPAH. Females are more likely to be affected, and most cases typically manifest in the fourth decade of life [5,6].

The common presenting symptoms of PAH include breathlessness on exertion, fatigue, chest pain, and dry cough; it may also present with non specific symptoms such as generalised weakness. The most commonly used method for diagnosing PAH is transthoracic ECG [7]. Right heart catheterisation is an invasive procedure that is considered the gold standard for diagnosing IPAH [7].

There is no definitive cure for IPAH, treatment options include a variety of medications, such as calcium channel blockers, prostanoids, endothelin receptor antagonists, phosphodiesterase 5 inhibitors, soluble guanylate cyclase stimulators like riociguat, selective prostacyclin IP receptor agonists such as selexipag, and lung transplantation in advanced cases [8].

A similar case report by Nugraha IW et al., highlighted the rare and serious complication of haemoptysis in a young female with PAH [9].

In the present case, a 24-year-old male with no co-morbidities and only complaints of streaky haemoptysis over the previous five years, for which he had been receiving symptomatic treatment, was diagnosed with IPAH following a thorough inquiry and extensive work-up.

While women are more likely to develop this condition and most cases typically occur in the fourth decade of life, the patient in present case was a young man (24 years old), which is uncommon.

In a study from India, however, the mean age at diagnosis of IPAH was found to be lower in men (25±7.6 years) than in women (28.5±12.9 years) [10]. The reason for this variation is not known.

Patients with IPAH generally present with exertional dyspnoea, chest discomfort, and cough. However, in our case, the primary complaint was streaky haemoptysis, which persisted for five years before dyspnoea developed. It is rare for a patient with IPAH to present with only haemoptysis and no other complaints.

Haemoptysis is a concerning condition that occurs infrequently in people with pulmonary arterial hypertension. Reports indicate that it is a rare and critical stage complication in patients already diagnosed with PAH. The occurrence of haemoptysis as the chief presenting complaint in PAH patients is not well-documented and is frequently underreported. The pathogenesis of haemoptysis in PAH remains unclear, and the mortality associated with haemoptysis in PAH is influenced by multiple factors. Some studies suggest that individuals with PAH and congenital heart disease have a better prognosis regarding haemoptysis than those with other categories of PAH. However, the fundamental mechanisms behind this syndrome are still not fully understood [10,11].

In the present case, the unique presentation and lack of suspicion led to a delay in the diagnosis of IPAH. After ruling out all other causes of pulmonary hypertension, we diagnosed the patient with IPAH {World Health Organisation (WHO) Functional Class 3} [1]. The patient reported no further episodes of haemoptysis and is demonstrating good symptom control.

CONCLUSION(S)

The present case underscores the importance of evaluating uncommon symptoms, such as haemoptysis, which can sometimes be the sole presenting feature of IPAH and highlights the critical need to consider IPAH, even when patients present with atypical symptoms. Clinicians should maintain a high index of suspicion for IPAH, regardless of gender. It is essential to diagnose and initiate prompt treatment of IPAH as early as possible. With advancing diagnostic methods and the availability of newer medications—including calcium channel blockers, prostanoids, endothelin receptor antagonists, phosphodiesterase-5 inhibitors, soluble guanylate cyclase stimulators like riociguat, and selective prostacyclin IP receptor agonists such as selexipag—there is potential to reduce mortality, especially when the disease is suspected and identified early.

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