DOI: https://doi.org/10.4038/ajim.v5i1.359

CASE REPORT

Asian J Intern Med. 2026 Jan-Feb; 5(1)

Hereditary haemorrhagic telangiectasia: an unusual cause for a quiescent pulmonary nodule

Sobika S[1]*, Suganthan N[1], Kumanan T[1], Sriluxayini M[1], Gerald S[1]

Abstract

Hereditary haemorrhagic telangiectasia (HHT) is a rare autosomal dominant disorder with mucocutaneous telangiectasias and arteriovenous malformations (AVM). We report an asymptomatic patient with a pulmonary AVM (PAVM) found on routine imaging. He later disclosed recurrent childhood epistaxis and a family history suggestive of HHT. Imaging studies confirmed a large PAVM without other organ involvement. Early diagnosis and transcatheter embolisation helped prevent severe complications like stroke and haemorrhage. This case highlights the importance of exclusion of HHT as an important cause of an unexplained pulmonary shadow.

Keywords: hereditary haemorrhagic telangiectasia, pulmonary arteriovenous malformation, Curaçao criteria, bubble echocardiogram

Introduction

Hereditary haemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu disease, is a rare autosomal dominant disorder of the fibrovascular tissue caused by mutations in ACVRL1, ENG, or SMAD4, which affect TGF-β signaling and vascular development.(1) Diagnosis is based on the Curaçao criteria, which include spontaneous recurrent epistaxis, multiple telangiectasias, visceral involvement, and a family history.(2) The most common symptom is epistaxis, affecting 90–95% of patients, usually beginning in childhood.(3) Other features include telangiectasias on the skin, oral mucosa, nail beds, gastrointestinal bleeding, and iron deficiency anaemia. Silent visceral lesions in the lungs, liver, stomach, or bowel may go unnoticed until complications arise.(4) The most frequent pulmonary manifestation is pulmonary arteriovenous malformation (PAVM), which can lead to stroke, septic emboli, or pulmonary haemorrhage.(1) Transthoracic contrast echocardiography (TTCE) is the preferred screening tool due to its high sensitivity, while contrast-enhanced pulmonary angiography remains the gold standard for diagnosis.(5) Management includes transcatheter embolisation to prevent embolic events, haemorrhage, and hypoxaemia.(1)

Case presentation

A 27-year-old man was evaluated for an incidental lung nodule found on a chest X ray during a medical checkup. He denied any symptoms related to this heterogeneous lung shadow. The patient had no haemoptysis, anorexia or weight loss. He had a normal exercise tolerance. On examination he had an average body built, plethoric with mild cyanosis of lip

*Correspondence:

Shanmugavadivel Sobika
Registrar in general medicine
Teaching hospital Jaffna, Sri Lanka
E-mail: sobikasiva1993@gmail.com
Phone: 077518317

Full list of author information is available at the end of the article

The Official Journal of
Sri Lanka College of Internal Medicine

and tongue. He had tiny telangiectasia on his tongue. He did not have skin telangiectasia or lymphadenopathy. A grade 2 clubbing was noted over the fingers and toes. His blood pressure was 120/80 mmHg with a pulse rate of 80 beats per minute. Lung and cardiac examination findings were unremarkable. He had an oxygen saturation of 90% on ambient air. He didn't have ankle oedema or precordial signs suggestive of pulmonary hypertension. With the clinical suspicion of hereditary haemorrhagic telangiectasia, on direct enquiry he revealed a history of episodic self-limiting epistaxis from childhood. He also had nail bed bleeding in the past which was not evaluated. No other bleeding sites were identified. He denied any symptoms suggestive of anaemia. No other significant past medical or surgical history were noted. His mother and elder brother were also having epistaxis however they were not evaluated. He never smoked or abused alcohol.

Arterial blood gas analysis showed: pH of 7.393, P_AO_2 of 71mmhg, and $PaCO_2$ of 29.2mmhg and SpO2 of 90%. A summary of investigations are given in Table 1.

His liver function including coagulations profile, renal function and inflammatory markers were normal. His chest x-ray showed a heterogeneous mass like lesion in the right lower zone which was suspected to be an arteriovenous malformation with the given history and examination findings (Figure 1A).

Computed tomography pulmonary angiogram (CTPA) and Computed tomography chest confirmed an arteriovenous malformation in the right poster basal and right lateral basal segments, measuring 3.0 cm (craniocaudal) × 3.8 cm (transverse) × 3.2 cm (anteroposterior) (Figure 1B). A large feeding pulmonary artery arose from the right interlobar pulmonary artery and drained to the right inferior pulmonary vein. There were no similar lesions in the rest of the lung.

Cardiac function was also assessed with a 2D Echocardiogram which showed satisfactory biventricular function without evidence of pulmonary hypertension. Bubble echocardiogram was done which demonstrated air bubbles in the right ventricle and atrium immediately after injection. At the same time there were no bubbles observed in the left chambers. After a few seconds (3- 5 seconds) bubbles appeared in the left chamber as well. This study concluded that there was no intracardiac shunt and that there is a high possibility of arteriovenous pulmonary shunting.

Table 1. Haematological investigations during ward admission

Investigation	Patient values	Reference range
WBC ($x10^9$/L)	8.11	4.00-11.00
Neutrophils ($x10^9$/L)	3.70	2.00-7.00
Lymphocytes ($x10^9$/L)	3.03	0.80-4.00
Eosinophils ($x10^9$/L)	0.84	0.02-0.50
Basophils ($x10^9$/L)	0.05	0.00-0.10
Monocytes ($x10^9$/L)	0.49	0.12-1.20
Haemoglobin (g/dL)	17.0	12.0-16.0
RBC (10^{12}/L)	5.60	4.00-5.50
HCT (%)	51.9	40.0-54.0
MCV (fL)	87.6	80.0-100.0
MCH (pg)	28.6	27.0-34.0
MCHC (g/dL)	32.8	32.0-36.0
RDW (%)	15.2	11.0-16.0
Platelets ($x10^9$/L)	202	150-450

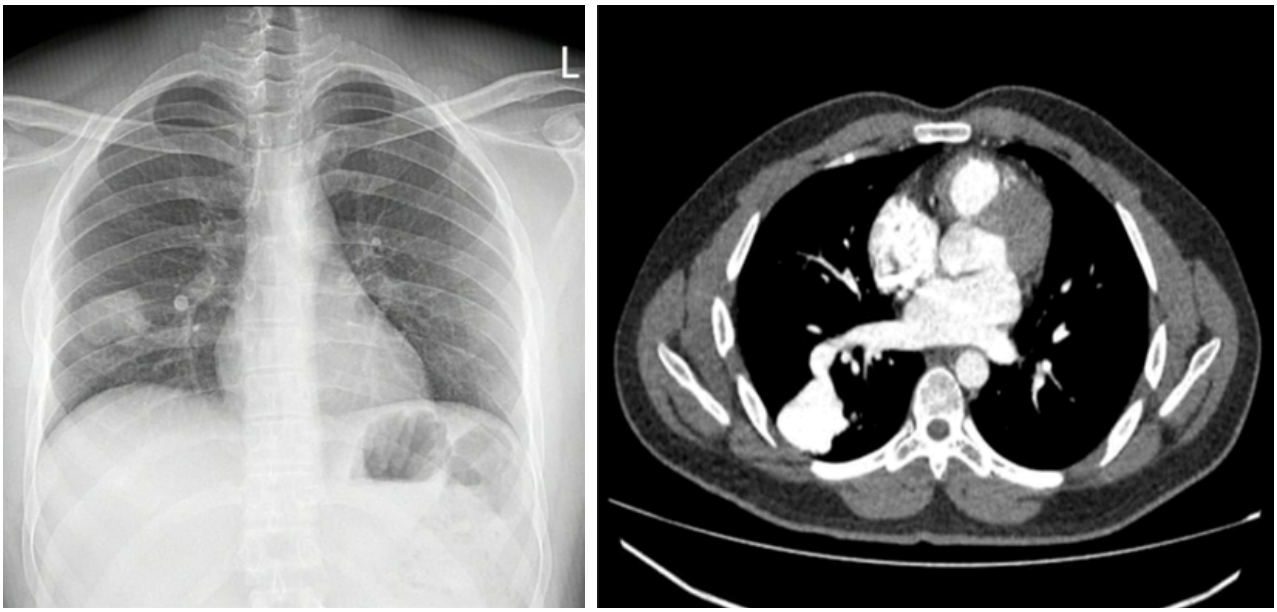

Figure 1. Chest x-ray shows a heterogenous mass like lesion in right lower zone and CTPA shows a pulmonary arteriovenous malformation

There were no upper gastrointestinal tract telangiectasias in upper esophagogastroduodenoscopy study. He was referred to the interventional radiology team for embolisation or coiling of arteriovenous malformation.

Discussion

Hereditary haemorrhagic telangiectasia (HHT), or Osler-Weber-Rendu disease, is a rare autosomal dominant vascular disorder caused by mutations in ACVRL1, ENG, or SMAD4, affecting TGF-β–mediated vessel development.(1,6) Diagnosis is based on Curaçao criteria, which includes recurrent epistaxis, multiple telangiectasias, visceral involvement, and positive family history.(2)

Our patient fulfilled all criteria, like recurrent epistaxis, tongue telangiectasias, pulmonary arteriovenous malformation (PAVM), and family history of epistaxis which defined the definitive diagnosis of HHT. Like most of the PAVM patients, he was asymptomatic but had mild cyanosis, tiny telangiectasia on the tongue, and low oxygen saturation (90%) and clubbing suggesting right-to-left shunting. Laboratory results were normal except for mild polycythaemia, likely from chronic hypoxaemia. Significant proportion of the patients also have an associated iron deficiency anaemia due to associated gastrointestinal (GI) telangiectasias with bleeding and significant epistaxis.(7,8) But our patient had polycythaemia without iron deficiency suggestive of minimal GI tract involvement with prominent pulmonary involvement. A chest x-ray and CT pulmonary angiogram confirmed a large right lower lobe PAVM. Transthoracic contrast echocardiography also showed delayed left heart bubble appearance, confirming extracardiac shunt with normal cardiac function without pulmonary hypertension. Endoscopy excluded gastrointestinal telangiectasias. Although genetic testing was advised for a definitive diagnosis, it could not be pursued due to financial constraints, which is a recognised barrier in the management of HHT patients globally.

PAVM is the most common pulmonary manifestation of HHT. It may remain silent until complications such as stroke, brain abscess, or haemorrhage occur. Prevalence and incidence of PAVM rupture is rare (2.7% and 0.16%, respectively). This was demonstrated in a study in the United States of America which included only 801 patients from 2 July 1996 and 22 July 2021.(9) Standard treatment for PAVM is transcatheter embolisation to prevent embolic and haemorrhagic complications while improving oxygenation.(1) Our patient was referred for the procedure.

This case underscores the importance of detailed history and examination in identifying HHT, as silent

but potentially life-threatening vascular lesions may be present. Early recognition and intervention can prevent serious complications.

Conclusion

Hereditary haemorrhagic telangiectasia (HHT) is a rare but potentially serious genetic disorder usually presenting with recurrent overt bleeding. However, these bleeding episodes are trivial and often overlooked. This case illustrates the importance of taking a detailed past medical history and family history, related to bleeding diathesis in particular, in evaluating an unexplained pulmonary nodule which might be a PAVM which could end up with devastating complications at a later stage if not intervened appropriately.

Author details

[1]Teaching Hospital Jaffna, Sri Lanka

Declarations

Authors' Contributions:
SS was the registrar in medicine who was responsible for the patient. Other authors supervised the management. The manuscript was prepared by all authors involved, collectively. All authors participated in manuscript revision, agreed to submit the manuscript and approved the final version of the manuscript.

Data Accessibility:
All data supporting this report are contained within the manuscript.

Funding: No specific funding was received.

Conflicts of interest:
The authors declare that they have no conflicts of interest.

Acknowledgements:
The authors would like to acknowledge the contributions made by the other subspecialties involved in managing the patient.

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Received: 12 Aug 2025 **Accepted:** 03 Jan 2026