Int. j. adv. multidisc. res. stud. 2025; 5(6):549-552

Received: 24-09-2025
Accepted: 04-11-2025

International Journal of Advanced Multidisciplinary Research and Studies

ISSN: 2583-049X

Case Report: Multisystemic AVMs on Cross-Sectional Imaging: Uncovering Hereditary Haemorrhagic Telangiectasia in an Indian Male

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Abstract

Hereditary Haemorrhagic Telangiectasia (HHT), (also termed as Osler-Weber-Rendu syndrome) is a genetic vascular condition marked by telangiectatic vascular lesions of the skin and mucous membranes, accompanied by arteriovenous malformations involving visceral organs. We present a case of HHT in a patient who exhibited recurrent nose bleeds and haemoptysis, cutaneous telangiectasias, pulmonary arteriovenous malformations, and hepatic vascular involvement. Diagnosis was confirmed according to the Curaçao criteria and further substantiated by imaging, highlighting the need for thorough organ evaluation and multidisciplinary management in HHT patients, even when neurological manifestations are not present.

Keywords: Hereditary Haemorrhagic Telangiectasia, Osler-Weber-Rendu Disease, Pulmonary AVMs, Hepatic Telangiectasias, Epistaxis, Embolotherapy

Introduction

Hereditary Haemorrhagic Telangiectasia (HHT) is an inherited autosomal dominant condition characterized by telangiectatic lesions of the skin and mucous membranes along with arteriovenous malformations affecting internal organs. Its prevalence is estimated to be 1 in 5,000–10,000, but the condition is frequently underdiagnosed owing to mild or asymptomatic presentations [1,2].

HHT develops due to genetic alterations affecting the transforming growth factor-beta (TGF-β) signalling pathway, with ENG and ACVRL1 mutations being the most prevalent, while SMAD4 variants occur less often. These genes are essential for angiogenesis and vascular stability, and their disruption causes endothelial dysfunction, giving rise to fragile, dilated vessels lacking intervening capillaries—thereby predisposing patients to haemorrhage and arteriovenous shunting [3]. Genotype–phenotype associations have also been described: ENG mutations are more often associated with pulmonary and cerebral AVMs (HHT type 1), while ACVRL1 mutations are linked to hepatic vascular malformations (HHT type 2) [4, 5].

Diagnosis is based on the Curaçao criteria, which include: (1) recurrent epistaxis, (2) multiple telangiectasias at typical sites, (3) visceral arteriovenous malformations, and (4) a first-degree relative with HHT. The presence of three or more criteria establishes a definite diagnosis, while two suggest possible HHT [6]. Genetic testing provides confirmatory evidence and aids in family screening.

Visceral arteriovenous malformations are the main contributors to morbidity in HHT. Pulmonary AVMs are identified in roughly one-third to one-half of HHT cases and give rise to right-to-left shunts, resulting in hypoxemia and permitting emboli to bypass the pulmonary circulation, with potential outcomes including stroke or brain abscess [7]. Hepatic AVMs, affecting up to 78% of patients, most notably those with ACVRL1 variants commonly remain subclinical but can manifest as high-output cardiac failure, portal hypertension, or ischemic biliary injury [5, 8]. Mucocutaneous telangiectasias, typically affecting the lips, tongue, and hands, often constitute the earliest visible manifestation.

This case describes a patient with classic cutaneous telangiectasia and visceral involvement (pulmonary and hepatic AVMs) but without cerebral AVM. It highlights the clinical variability of HHT and reinforces the importance of systematic screening,

genetic evaluation when available, and multidisciplinary management to prevent severe complications.

Clinical Background

A 32-year-old man presented to the outpatient clinic with a history of recurrent spontaneous nosebleeds and haemoptysis since adolescence, which had gradually increased in both frequency and duration over recent years. The episodes were typically self-limiting. The patient also reported intermittent shortness of breath on exertion but denied chest pain, palpitations, or syncope. There was no history of cyanosis or neurological symptoms such as seizure, headache, or focal deficits. Family history of similar complaints was noted in his mother and daughter (Figure 1a).

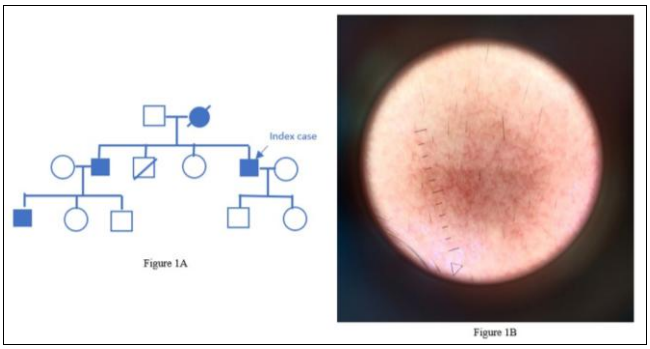

Fig 1a: Pedigree chart showing our case as the index case, with similar history in patient's mother and brother; **Fig 1b:** Blanching cutaneous telangiectasias confirmed by contact dermoscopy

General physical examination revealed multiple discrete, blanching telangiectasia over face and lips, confirmed via contact dermoscopy (Figure 1b). Vital signs were within normal limit, with resting oxygen saturation of 97% on room air. Cardiovascular and respiratory system examination was unremarkable, with no audible murmur or sign of right heart strain. Abdominal examination was soft and non-tender, with no hepatosplenomegaly or ascites. Neurological examination was normal.

Imaging Findings

Contrast-enhanced Computed Tomography (CT) of chest showed few, well-defined lobulated contrast-filled outpouchings located in left lower lobe and right upper lobe (Figure 2a). These lesions appeared as nodular vascular structure with feeding arteries measuring between 3–6 mm and early draining pulmonary veins, consistent with pulmonary arteriovenous malformation (PAVM) (Figure 2b). 3D reconstruction of CECT images better delineate arterial feeders and early draining pulmonary veins, with outpouchings (Figure 2c). No evidence of associated haemorrhage, infarction, or parenchymal consolidation was observed.

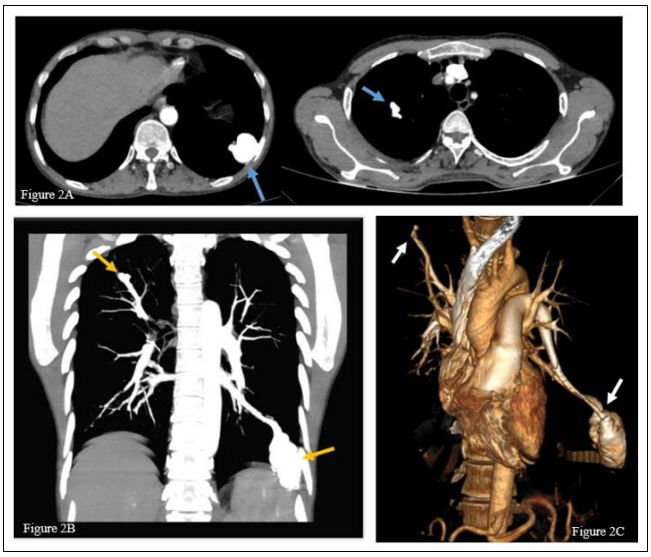

Fig 2a: Arterial phase CECT images of Chest in axial plane show well-defined, contrast filled outpouchings in left lower lobe and right upper lobe (Blue solid arrows); **Fig 2b:** Coronal MIP arterial phase CECT images of chest show communication of contrast-filled outpouchings with feeding arteries and early draining pulmonary veins, suggestive of Pulmonary Arteriovenous Malformations (PAVMs) (Orange solid arrows); **Fig 2c:** 3D reconstruction of CECT images better demonstrate presence of arterial feeders and draining veins in PAVMs (white solid arrows)

Contrast-enhanced CT of abdomen showed heterogeneous arterial phase enhancement of liver with multiple small telangiectatic vessel, involving hepatic lobes. These appeared as punctate enhancing foci and early filling of hepatic veins during arterial phase, suggestive of intrahepatic arteriovenous shunting (Figure 3). There was no biliary dilation, ascites, or evidence of portal hypertension.

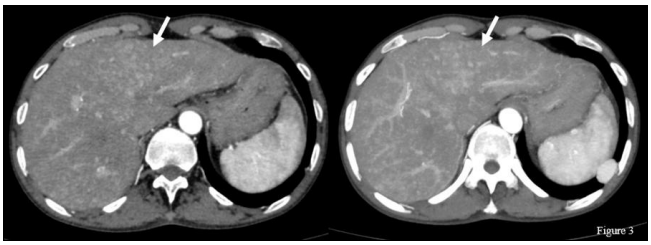

Fig 3: Arterial phase CECT images of abdomen in axial plane show multiple punctate arterial phase enhancing foci in both hepatic lobes (white solid arrows) with early filling of hepatic veins, suggestive of hepatic telangiectasias

Magnetic resonance imaging (MRI) of brain was performed for evaluation of any cerebral AVM. The study revealed normal brain parenchyma with no evidence of AVM, cavernous malformation, or vascular anomaly on contrast-enhanced sequences and MR angiography.

These imaging findings supported diagnosis of multisystemic HHT with involvement of pulmonary and hepatic vascular beds, in absence of cerebral disease. This patient is being evaluated by Interventional Radiology team for embolization of PAVM.

Discussion

Hereditary Haemorrhagic Telangiectasia (HHT) is a multisystem vascular dysplasia with variable clinical severity. The present case illustrates classical manifestations, including mucocutaneous telangiectasia, pulmonary AVMs, and hepatic vascular anomalies, but notably lacked cerebral AVMs, underscoring disease heterogeneity even in clinically definite cases.

In India, HHT remains under-reported and under-diagnosed, despite a large population base. Limited awareness, lack of genetic testing, and nonspecific symptoms such as anaemia or epistaxis often attributed to common causes contribute to missed diagnoses. Fewer than a dozen well-documented Indian cases exist in indexed literature, most without molecular confirmation [10].

Genetic testing is the gold standard for confirming HHT and identifying familial mutations, but it is often inaccessible in low-resource settings. Diagnosis in this case was based on clinical features and imaging findings fulfilling the internationally accepted Curaçao criteria.

Cutaneous telangiectasias, although benign, are important diagnostic clues, typically involving the face, lips, oral mucosa, and hands. Their presence is crucial for fulfilling the Curaçao criteria and prompting systemic evaluation [2, 6].

Pulmonary AVMs (PAVMs) are a major source of morbidity due to right-to-left shunting. In this patient, chest CT confirmed PAVMs, which occur in 30–50% of individuals with HHT, particularly in those with ENG mutations (HHT type 1) [3, 7]. These lesions bypass the pulmonary capillary filter, allowing unfiltered emboli or pathogens to enter systemic circulation, causing stroke, brain abscess, or paradoxical embolism [4]. Transcatheter embolization is the preferred therapy for PAVMs with feeding arteries ≥2–3 mm. It improves oxygenation and reduces systemic complications, though recurrence may occur, necessitating long-term follow-up [9].

Hepatic telangiectasias, although asymptomatic here, further support the multisystem nature of HHT. Hepatic involvement is common, particularly with ACVRL1 mutations (HHT type 2), and may affect up to 78% of patients [5, 8]. While often silent, imaging with Doppler ultrasound, contrast CT, or MRI is essential for detection and monitoring for any complications. Conservative management was appropriate in this case given absence of symptoms.

The absence of cerebral AVMs is noteworthy but not uncommon. Brain AVMs occur in 10–23% of HHT patients, more often in ENG mutation carriers, and may remain silent or present catastrophically with haemorrhage, seizures, or neurologic deficits. Guidelines recommend at least one screening MRI in all patients with suspected or definite HHT to aid risk stratification [1]. Normal neuroimaging in this case eliminated immediate intervention but highlighted the importance of baseline evaluation.

This patient fulfilled Curaçao criteria, supported by recurrent epistaxis, telangiectasia, visceral AVMs, and characteristic imaging findings. While these criteria remain fundamental, molecular testing provides definitive confirmation, aids familial screening, and may predict organ involvement [2].

HHT requires lifelong, organ-specific surveillance, as manifestations evolve over time. A multidisciplinary team involving radiology, interventional specialists, otorhinolaryngology, hepatology, genetics, and primary care is essential. Patient and family education regarding complications and the importance of routine follow-up is equally critical for preventing delayed detection of treatable lesions.

Conclusion

HHT is a multisystem vascular disorder that requires high index of suspicion and systematic screening, to prevent serious complication. This case highlights classical involvement of pulmonary and hepatic circulation, alongside cutaneous findings, and pivotal role of imaging in diagnosis and management. Embolotherapy remains effective for symptomatic pulmonary AVMs, while hepatic telangiectasia warrant ongoing observation. Early diagnosis and coordinated, lifelong surveillance are essential for improving outcomes in patients with HHT.

Teaching Points

1. The history of the patient suggesting recurrent, spontaneous epistaxis since adolescence highlights the fact that they are often the first and most common clinical manifestation, and when coupled with family history, should immediately prompt for evaluation of any systemic disease.
2. The patient's pedigree chart showing similar complaints in his mother and daughter tells about the autosomal dominant inheritance pattern of HHT. It demonstrates the requirement of screening in first-degree relatives with clinical and imaging evaluation, even if they are asymptomatic.
3. In low-resource settings like India, genetic testing for cases like HHT is often not available due to high cost and limited availability. Hence, diagnosis is based mainly on Curaçao criteria which is supported by clinical findings and cross-sectional imaging, This shows timely recognition and management despite limitation of molecular diagnosis.

MCQ questions

1. In Hereditary Hemorrhagic Telangiectasia (HHT), the pathophysiology for the formation of visceral arteriovenous malformations is because of:

A. Defective elastin synthesis and arrangement in vascular walls
B. Mutations involving endothelial TGF-β signaling pathway
C. Antibody mediated endothelial damage
D. Acquired liver cirrhosis due to vascular remodeling

Answer. B. Mutations involving endothelial TGF-β signaling pathway

2. Which of the following is NOT required in Curaçao criteria for diagnosing HHT?

A. Family history in a first-degree relative
B. Recurrent spontaneous epistaxis
C. Multiple mucocutaneous telangiectasias
D. Congenital bilateral cataracts

Answer. D. Congenital bilateral cataracts.

3. Which of the following complications of pulmonary AVMs in HHT is most directly related to right-to-left shunting?

A. Pulmonary hypertension
B. Hypoxemia and paradoxical embolic stroke
C. Hemoptysis due to bronchial artery rupture
D. Portal hypertension

Answer: B. Hypoxemia and paradoxical embolic stroke

Abbreviations

HHT	Hereditary Haemorrhagic Telangiectasia
CT	Computed Tomography
MRI	Magnetic Resonance Imaging
PAVM	Pulmonary Arteriovenous Malformation
AVM	Arteriovenous Malformation

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