# RECURRENT HEMORRHAGIC MANIFESTATIONS IN A PATIENT WITH HEREDITARY HEMORRHAGIC TELANGIECTASIA AND PULMONARY ARTERIOVENOUS MALFORMATION: A CASE REPORT

**Viljeena Wilson[1], Christy Binu[1]**
[1]Department of Clinical Pharmacy, Mother Hospital, Olari, Kerala, India
**CORRESPONDANCE**
Clinical Pharmacist, Department Of Clinical Pharmacy, Mother Hospital, Olari, thrissur 680012, Kerala, India

## ABSTRACT

Hereditary hemorrhagic telangiectasia (HHT) is a rare autosomal dominant vascular disorder characterized by mucocutaneous telangiectasias and visceral arteriovenous malformations (AVMs), leading to recurrent bleeding and multisystem complications. Pulmonary arteriovenous malformations (PAVMs) are a major cause of morbidity due to hemoptysis, hypoxemia, and paradoxical embolic events.

We report the case of a 63-year-old man with a known diagnosis of HHT and a history of endovascular embolization of a pulmonary AVM who presented with acute epistaxis, oral bleeding, and massive hemoptysis. Laboratory evaluation revealed anemia with normal platelet count and coagulation profile. Computed tomography pulmonary angiography demonstrated previously embolized AVM in the left upper lobe with surrounding ground-glass opacities suggestive of pulmonary hemorrhage.

The patient was managed conservatively with antifibrinolytic therapy, blood transfusion, antimicrobial treatment, blood pressure control, and supportive care, resulting in clinical stabilization without recurrence of massive bleeding during hospitalization.

This case highlights the persistent risk of hemorrhagic complications in patients with HHT despite prior definitive treatment and emphasizes the need for lifelong surveillance and multidisciplinary management to reduce morbidity and prevent life-threatening events.

**KEYWORDS:** Hereditary hemorrhagic telangiectasia; Pulmonary arteriovenous malformation; Hemoptysis; Epistaxis

## INTRODUCTION

Hereditary hemorrhagic telangiectasia (HHT), also known as Osler–Weber–Rendu syndrome, is a rare inherited vascular disorder resulting from abnormalities in angiogenesis and vascular remodeling [1]. It follows an autosomal dominant inheritance pattern with variable penetrance and age-dependent clinical expression. The estimated prevalence ranges between 1 in 5,000 and 1 in 10,000 individuals, though the condition is frequently underdiagnosed, particularly in resource-limited settings [2,3].

HHT is characterized by the presence of mucocutaneous telangiectasias and visceral arteriovenous malformations (AVMs), which arise due to direct artery-to-vein connections without an intervening capillary

bed. These fragile vascular structures predispose patients to recurrent bleeding and systemic complications. Mutations affecting the transforming growth factor-β signaling pathway, including ENG, ACVRL1, and SMAD4 genes, play a key role in disease pathogenesis [4].

Clinically, recurrent spontaneous epistaxis is the most common manifestation and often represents the earliest symptom. With disease progression, telangiectasias may appear on the lips, oral cavity, face, and extremities. Visceral AVMs involving the lungs, brain, liver, and gastrointestinal tract contribute significantly to morbidity and mortality [5]. Pulmonary AVMs are particularly important due to their association with hypoxemia, hemoptysis, paradoxical embolism, ischemic stroke, and brain abscess formation [6].

Diagnosis is primarily clinical and based on the Curaçao criteria. Endovascular embolization is the preferred treatment for pulmonary AVMs; however, recurrence, recanalization, and development of new lesions may occur, necessitating lifelong follow-up [7]. This report describes a patient with HHT who developed recurrent hemorrhagic manifestations despite prior pulmonary AVM embolization.

## CASE PRESENTATION

A 63-year-old male presented to the emergency department with sudden-onset epistaxis and oral bleeding accompanied by profuse hemoptysis lasting approximately 30 minutes. The bleeding was abrupt in onset, with expectoration of large volumes of fresh blood. There was no history of trauma, fever, chest pain, or recent invasive procedures.

The patient had a known diagnosis of hereditary hemorrhagic telangiectasia established several years earlier based on recurrent epistaxis, mucocutaneous telangiectasias, and visceral involvement. His medical history was significant for pulmonary arteriovenous malformation treated with endovascular embolization in 2017, septoplasty for nasal telangiectasias in 2015, splenic abscess and thyroid abscess, and a cerebral abscess involving the left frontal lobe complicated by seizures in 2020. He also had comorbid hypertension and type 2 diabetes mellitus.

On examination, the patient was conscious and oriented, with stable vital signs and no evidence of hypoxia. Active nasal and oral bleeding was noted. Telangiectasias were visible on the lips and oral mucosa. Respiratory examination revealed bilateral inspiratory crepitations, more pronounced over the left lung fields. Cardiovascular and neurological examinations were unremarkable.

Laboratory investigations revealed anemia with a hemoglobin level of 9.3 g/dL, which declined during hospitalization. Leukocyte count was elevated (12,700 cells/mm$^3$), and erythrocyte sedimentation rate was markedly raised (99 mm/hr). Platelet count and coagulation parameters, including international normalized ratio (1.10), were within normal limits.

Computed tomography of the brain showed chronic gliotic changes without acute hemorrhage. Computed tomography pulmonary angiography demonstrated evidence of previously embolized pulmonary AVM in the left upper lobe with surrounding ground-glass opacities, consistent with pulmonary hemorrhage.

The patient was managed conservatively with intravenous tranexamic acid, piperacillin–tazobactam, blood pressure control using labetalol, nasal hemostatic agents, and supportive care. One unit of packed red blood cells was transfused. No further episodes of massive hemoptysis occurred, and the patient was discharged in stable condition with advice for long-term follow-up.

## DISCUSSION

Pulmonary arteriovenous malformations are a major contributor to morbidity in patients with HHT and may lead to life-threatening hemorrhagic and neurological complications. Although endovascular embolization significantly reduces the risk of adverse outcomes, it does not eliminate the possibility of future bleeding [8].

In the present case, recurrent hemoptysis occurred several years after embolization, likely due to vascular fragility or recanalization rather than acute AVM rupture. Ground-glass opacities surrounding the embolized AVM suggested alveolar hemorrhage, highlighting the complex pathophysiology of bleeding in HHT.

Bleeding in HHT is structural in nature, rather than due to coagulation abnormalities, as evidenced by normal platelet counts and coagulation parameters in this patient. Inflammatory states and pulmonary infections may further exacerbate vascular instability and precipitate hemorrhagic events [9].

Conservative management with antifibrinolytic therapy, blood transfusion, and supportive care may be effective in hemodynamically stable patients without an identifiable bleeding source. Tranexamic acid has demonstrated benefit in reducing bleeding severity in HHT and is increasingly utilized in both acute and chronic management [10].

This case underscores the importance of lifelong surveillance and multidisciplinary care, particularly in patients with prior pulmonary AVMs and a history of systemic complications.

## CONCLUSION

Hereditary hemorrhagic telangiectasia is a lifelong multisystem disorder associated with recurrent hemorrhagic complications. This case demonstrates that patients remain at risk for significant bleeding events despite prior pulmonary AVM embolization. Early recognition, individualized conservative management during acute episodes, and coordinated multidisciplinary follow-up are essential to reduce morbidity and improve long-term outcomes.

## REFERENCES

1. Hermann R, Shovlin CL, Kasthuri RS, et al. Hereditary haemorrhagic telangiectasia. *Nat Rev Dis Primers*. 2025;11:1–20.
2. Ficany A, Del Alamo M, Bernabeu C, et al. Epistaxis prevention and treatment in HHT. *J Clin Med*. 2025;14:7724.
3. Kumar N, Marino MJ, Lal D. Innovations in HHT epistaxis management. *Front Allergy*. 2024;5:1456686.
4. El Sayed Ahmad Y, Kajal S, Halawi A. Managing epistaxis in HHT. *J Laryngol Otol*. 2024;138:847–857.
5. Beslow LA, White AJ, Krings T, et al. Screening for brain vascular malformations in HHT. *AJNR Am J Neuroradiol*. 2024;45:1177–1184.
6. Agarwal J, LaBranche J, Dhillon S, et al. Neurologic complications in HHT. *Can J Neurol Sci*. 2023;50:561–572.
7. Danesino C, Olivieri C. HHT genetics and diagnosis. *Pediatr Rep*. 2023;15:129–142.
8. L'Huillier R, Dupuis-Girod S, et al. Embolization outcomes in HHT. *CVIR Endovasc*. 2024;7:30.
9. Westermann CJ, de Vries V, et al. Multidisciplinary care in HHT. *Vasc Med*. 2023;28:153–165.
10. Schleupner MC, Röth A, et al. Iron deficiency in HHT. *Sci Rep*. 2025;15:28463.