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A CASE OF SPONTANEOUS HEMOTHORAX IN HEREDITARY HEMORRHAGIC TELANGIECTASIA(HHT)

Respiratory Medicine

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ABSTRACT

Introduction: Hereditary hemorrhagic telangiectasia is an inherited genetic disorder marked by irregularities in the formation of blood vessels in the skin, mucous membranes, and organs. Individuals with this condition typically display mucocutaneous telangiectasias and arteriovenous malformations in organs such as the lungs, liver, or central nervous system. **Case Report:** We describe a case of a 45-year-old female patient who presented with spontaneous hemothorax and was diagnosed with congenital hepatic arteriovenous malformation. Upon retrospective review, we identified the previously unnoticed components of the classic triad: recurrent epistaxis and a positive family history. This enabled us to establish a diagnosis of hereditary hemorrhagic telangiectasia. **Conclusion:** When encountering a patient with recurrent epistaxis, it is crucial to inquire about their family history and conduct a thorough examination for muco-cutaneous lesions. This approach facilitates early detection of hereditary hemorrhagic telangiectasia, allowing for diligent monitoring to prevent serious complications associated with cerebral and pulmonary arteriovenous malformations.

KEYWORDS

INTRODUCTION

The manifestations of hereditary hemorrhagic telangiectasia (HHT) were initially described in 1896 by Henri Jules Louis Rendu, who outlined the classical features of epistaxis, mucocutaneous telangiectasias, and familial inheritance.

Subsequently, William Osler (1900) and Frederick Parkes Weber (1907) provided detailed case descriptions, leading to the eponym "Rendu-Osler-Weber."

Early estimates placed the prevalence of HHT at 1 in 100,000, but recent studies suggest it is more common than previously thought. Vascular abnormalities in HHT range from small telangiectases to larger arteriovenous malformations (AVMs) involving direct arteriovenous connections and aneurysms. Pulmonary AVMs (PAVMs) are the most common source of clinical complications, affecting approximately 30-60% of patients. Gastrointestinal, hepatic, and cerebral involvement occurs in 11-40%, 8-16%, and 5-11% of HHT cases, respectively.

Epistaxis often precedes other symptoms in HHT and is frequently unrecognized as a sign of the condition.

Hemothorax in HHT cases is very rare presentation. If presents with Hemothorax presents as chest pain breathlessness.
Diagnostic criteria for HHT include:
(1) spontaneous recurrent epistaxis
(2) multiple telangiectasias
(3) visceral involvement and
(4) family history.

Diagnosis is classified as definite if three or more criteria are present, probable with two criteria, and unlikely with one or none.

Case Report

A 44-year-old female patient presented with a five-day history of breathing difficulty and chest pain exacerbated by lying on her left side or standing. She had been hospitalized a month earlier for severe epistaxis, which required laser photocoagulation. During examination, her vitals were stable while lying or sitting, but her oxygen saturation dropped to 88% when standing. Physical examination revealed dullness on percussion and absence of breath sounds over the left hemithorax.

Laboratory investigations showed a total leukocyte count of 12,320, hemoglobin level of 9.4 g/dL, and an elevated ESR of 56 mm/hour. Arterial blood gas analysis indicated hypoxemia with a PO2 of 70 mmHg on room air.

Chest X-ray revealed diffuse opacity in the left lower zone with blunting of the costo-phrenic angle, suggesting left-sided pleural effusion. Thoracentesis was performed, yielding grossly hemorrhagic pleural fluid for diagnostic and therapeutic purposes (Fig.1).

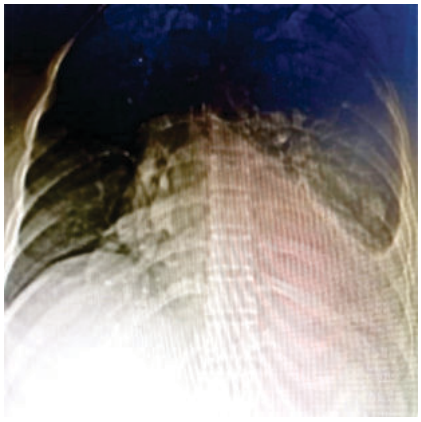

Fig.1: Chest X-ray done showed diffuse homogenous opacity in left lower zone with blunting of costo-phrenic angle Malignancy and tuberculosis was kept as working diagnosis .

However, within two days, the patient developed jaundice. An ultrasound of the abdomen and pelvis was performed, revealing multiple intrahepatic collaterals and several collaterals observed at the porto-hepatic junction (Fig.2).

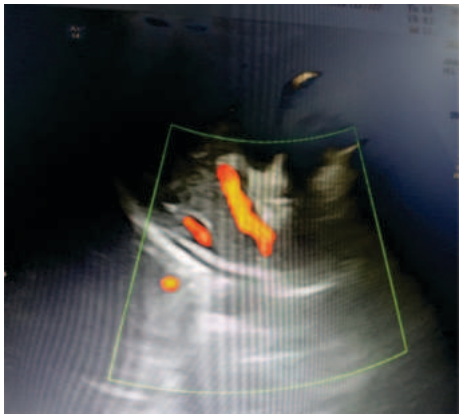

Fig.2:USG abdomen and pelvis done showed :Multiple intrahepatic collaterals with multiple collaterals seen at seen at portohepatic junction.

Despite considering the possibility of Budd-Chiari syndrome initially, the patient's clinical presentation did not align with this diagnosis. Therefore, a contrast-enhanced computed tomography (CECT) scan of the thorax and abdomen was performed. The imaging revealed moderate left hemothorax along with a pulmonary arteriovenous malformation (AVM) in the left lower lobe (Fig.4), characterized by focal vessel dilatation (aneurysm) with a risk of rupture (Fig.5).

The scan also showed a dilated and tortuous hepatic artery with multiple intrahepatic dilated and tortuous collaterals. Hepatic veins were noted to fill during the arterial phase, suggestive of congenital hepatic arteriovenous malformation (Fig.6).

Given the diagnosis of AVM, the focus shifted to the patient's previous admission for epistaxis. She reported 20 hospital visits in the past due to epistaxis, with four instances requiring laser photocoagulation. Regarding family history, she mentioned the neonatal death of her third child due to respiratory distress and jaundice. Her second child has a history of recurrent epistaxis.

Upon examination, the patient exhibited multiple mucocutaneous lesions throughout her body (Fig.3). These findings led to a clinical diagnosis of hereditary hemorrhagic telangiectasia (HHT) for the patient and a probable diagnosis for her son.

The patient underwent embolization of the pulmonary vessel, resulting in symptomatic improvement. Hemothorax was drained by ICD.

Fig.3: Mucocutaneous lesions noted on patients

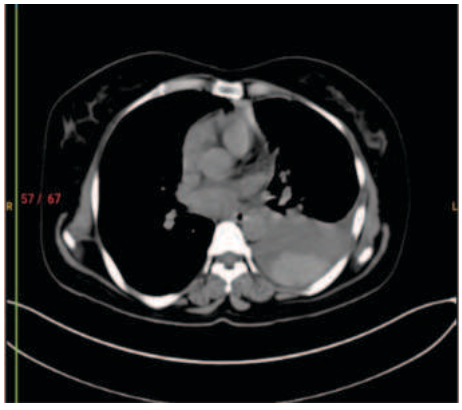

Fig.4: CECT thorax and abdomen done revealed moderate left hemothorax with pulmonary AVM in left lower lobe.

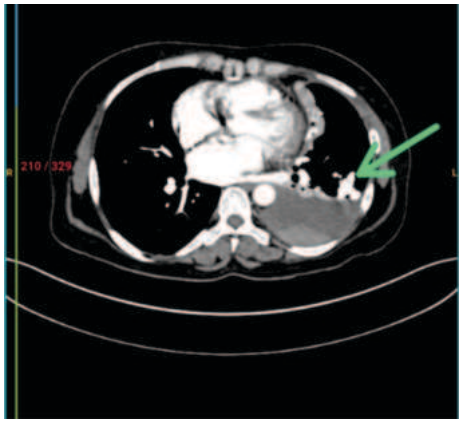

Fig.5: Focal dilatation of Pulmonary vessels (aneurysm)with possibility of rupture , dilated and tortous hepatic artery with multiple intrahepatic dilated and tortous collaterals

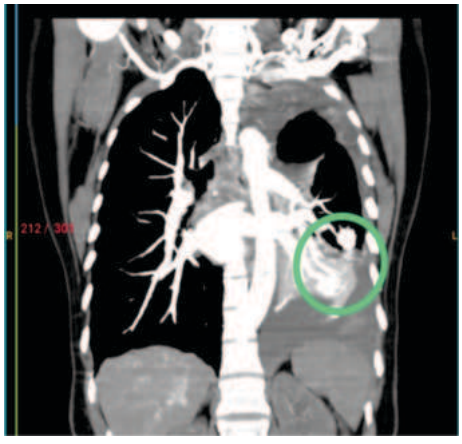

Fig.6: Hepatic veins opacifying in arterial phase suggestive of congenital hepatic artero venous malformation and aneurysm of vessels.

DISCUSSION

This case contributes significantly to our understanding of hereditary hemorrhagic telangiectasia (HHT). Our patient experienced her first episode of epistaxis at 14 years old, and her son at 10 years old. It is noted that approximately 50% of individuals with HHT report epistaxis by age 10, increasing to 80%-90% by age 21. Additionally, up to 95% of patients with HHT experience recurrent epistaxis, often due to the rupture of telangiectasias abundant in the nasal mucosa.

The patient's medical history revealed recurrent episodes of spontaneous nasal bleeding that went untreated by a physician. By age 16, 71% of those affected by HHT typically exhibit some symptoms, rising to over 90% by age 40.

Recent studies indicate that pulmonary arteriovenous malformations (PAVMs) occur in 50-60% of individuals with HHT. These lesions commonly appear in both lungs, particularly in the lower lobes, and are more frequently multiple rather than solitary, as observed in our patient.

Severe or potentially fatal hemorrhagic complications affect approximately 7% of HHT patients with PAVMs, such as intrabronchial or intrapleural rupture of PAVMs leading to hemoptysis or hemothorax, similar to our patient's presentation.

The diagnosis of HHT is based on the "Curaçao" criteria, which requires the presence of at least three out of four criteria for confirmation. Our patient met all four criteria.

In recent years, catheter ablation or embolization has become a primary treatment for PAVMs, although long-term follow-up studies are still lacking. New therapeutic approaches, including thalidomide and the monoclonal antibody Bevacizumab, have shown promise, but their efficacy requires further evaluation.

CONCLUSION

In summary, this study underscores the importance of heightened clinical awareness as an initial step in identifying patients with HHT, which is crucial for timely intervention.

Recognizing HHT promptly enables vigilant monitoring to mitigate potential severe complications associated with cerebral and pulmonary visceral arteriovenous malformations.

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