

Hereditary hemorrhagic telangiectasia with splenic abscess: a case report

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Abstract

Hereditary hemorrhagic telangiectasia (HHT) is a rare disorder. Although it commonly causes recurrent bleeding manifestations and severe iron deficiency anaemia, they may be overlooked. Splenic abscess is rarely seen in clinical practice in patients with HHT. This case report describes a patient who presented with epistaxis and severe anaemic symptoms and found to have a splenic abscess with underlying HHT.

Key words: hereditary hemorrhagic telangiectasia, splenic abscess, arteriovenous malformations, epistaxis

Introduction

Hereditary hemorrhagic telangiectasia (HHT; also called Osler-Weber-Rendu syndrome) is an autosomal dominant vascular disorder, with a variety of clinical manifestations that vary between relatives who have the same HHT pathogenic gene variant. The commonest clinical problems encountered are epistaxis, gastrointestinal bleeding, and iron deficiency anaemia, along with characteristic mucocutaneous telangiectasia. In addition, arteriovenous malformations (AVMs) frequently affect the pulmonary, hepatic, and/or cerebral circulations.⁽¹⁾ The majority of patients are unaware of their diagnosis of HHT and have not been diagnosed at the time of hospital admission. As a result, HHT has been subject to under-reporting.⁽²⁾

Case presentation

A 56-year-old woman with diabetes mellitus, recurrent nasal bleeding and iron deficiency anaemia, was admitted to Teaching Hospital - Jaffna for further evaluation of fever with a productive cough for one week. She also had shortness of breath on exertion, left pleuritic type chest pain and dull aching

hypochondrial pain for the same period. Her urine output and bowel habits were normal. She did not experience orthopnea or paroxysmal dyspnea. There was no contact history of tuberculosis. She denied a history of high-risk sexual behaviour. She did not have focal symptoms of malignancy or symptoms of autoimmune connective tissue diseases. Her father, brother and sister had suffered from recurrent nose bleeds. On further inquiry, she stated that she was treated for recurrent chest infections in the past.

On examination, she was febrile with a temperature of 102° F. She was severely pale. Her pulse rate and blood pressure were 108 bpm and 100/60 mmHg, respectively. Auscultation of the lungs revealed coarse crepitations with reduced breath sounds over the left lower zone. Examination of the abdomen revealed mild epigastric tenderness and the heart sounds were normal. There were no focal neurological signs or skin rashes.

Initial blood investigations revealed neutrophilic leukocytosis ($29.61 \times 10^9/L$), thrombocytosis ($499 \times 10^9/L$), microcytic hypochromic anaemia (haemoglobin 4.4 g/dL), elevated inflammatory markers (c-reactive protein-205.2 mg/L, erythrocyte sedimentation rate-135 mm/1st hour) with normal

renal and liver function tests. She had chest radiography which showed left lower lobe consolidation with mild pleural effusion and normal cardiac silhouette. She was commenced on intravenous(IV) fluids, IV ceftriaxone 2 g twice daily and oral doxycycline 100 mg twice daily for community acquired pneumonia. The Mantoux reading was negative and sputum studies could not be carried out since the sputum sample was inadequate. Transthoracic two-dimensional echocardiogram showed no evidence of infective endocarditis. Her urine full report and culture yielded no growth. Her blood picture showed severe iron deficiency anaemia and serum ferritin level was 23 ng/mL. Von Willebrand Factor (VWF) level, clotting profile and platelet function tests were normal. She had continuous fever and worsening of abdominal pain despite broad spectrum antibiotics so that an urgent contrast enhanced computed tomography of chest and abdomen was performed, which showed a hypoechoic lesion over the spleen ,measuring 5.0 x 5.0 x 6.0 cm, suggestive of splenic abscess with mild splenomegaly (figure1). Lung volume was preserved with left basal inflammatory shadow and a mild effusion suggestive of an ongoing inflammatory process with parapneumonic effusion. There was neither evidence of malignancy nor tuberculosis in this study.. Pus culture was sent, which isolated pseudomonas species. Antibody for melioidosis was negative. As her inflammatory markers did not show any improvement, she was started on intravenous piperacillin and tazobactam. A blood transfusion was also given. She was thoroughly investigated for the

cause of iron deficiency anaemia. Upper Gastrointestinal Endoscopy(UGIE), colonoscopy and Direct Fiber Optic Laryngoscopy (FOL) were performed where the latter revealed tiny multiple telangiectatic spots in the nasal cavity. She underwent capsular endoscopy, which also demonstrated multiple telangiectatic lesions throughout the small intestine(figure2). A diagnosis of HHT with splenic abscess and recurrent chest infections was made given the family history of nosebleeds. Upon recovery she was given health education regarding her condition and the need for proper screening and follow up.

Discussion

The combination of epistaxis, gastrointestinal bleeding, and iron deficiency anaemia associated with characteristic telangiectasia on the lips, oral mucosa, and fingertips has become firmly established as a medical entity.(3)

The classical constellation of findings underestimates potentially life-threatening aspects of HHT. In major series to date, at least one-half of HHT patients have pulmonary arteriovenous malformations (PAVMs), placing them at risk of early onset, preventable strokes; cerebral abscesses. Similarly hepatic AVMs affect approximately one-half of HHT patients and there is increasing concern about their implications. (1)

Most patients with HHT present with the symptoms

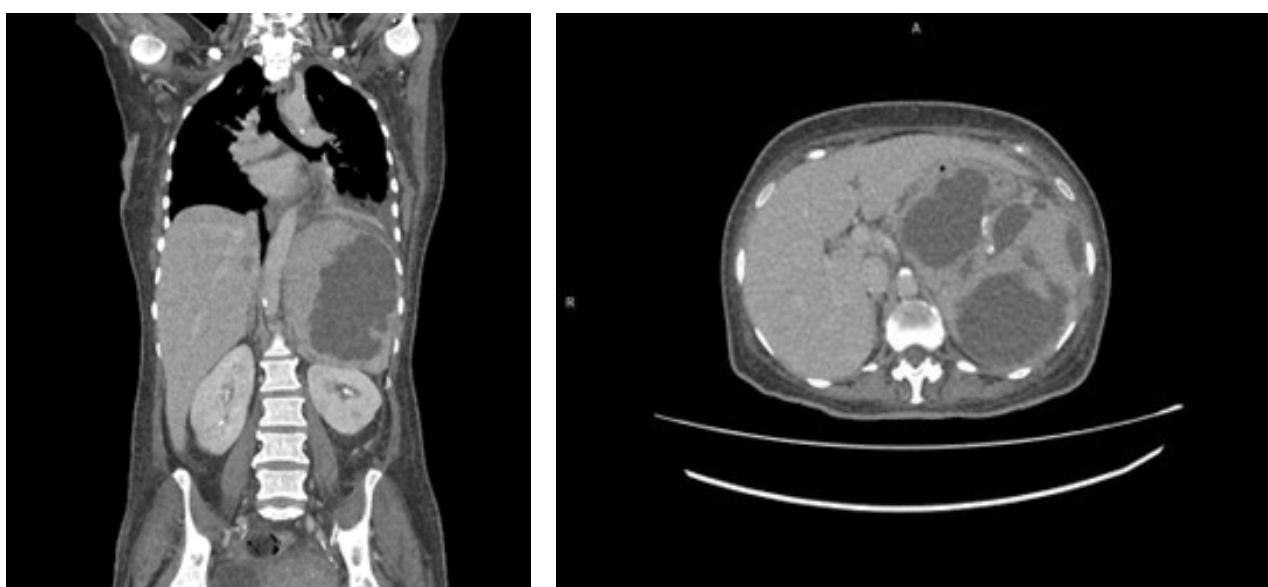


Figure 1 - Axial and coronal view of CECT abdomen show splenic abscess



Figure 2 - Capsular endoscopy shows multiple telangiectatic spots throughout the small intestine

and signs like epistaxis, mucocutaneous telangiectasia, and features of iron deficiency anaemia due to blood loss. However, some patients can have substantial symptoms, particularly attributable to severe recurrent nose bleeds and/or gastrointestinal bleeding, resulting in transfusion dependence, augmented when visceral arteriovenous malformations (AVMs) are present with resultant higher cardiac outputs. Epistaxis usually occurs early in the progression of the disease, often in childhood. Mucocutaneous and gastrointestinal telangiectasia develop later.(1) Our patient had recurrent nose bleeds, iron deficiency anaemia which required blood transfusion and gastrointestinal telangiectasia.

Spontaneous, recurrent epistaxis from telangiectatic patches of the nasal mucosa is the commonest presentation of HHT. Some patients experience no or minimal occasional episodes, but for the majority, recurrent and frequent epistaxis is a feature, with many patients experiencing daily bleeds.(4)

Recurrent gastrointestinal bleeding occurs in up to 33% of patients with HHT, often presenting as anaemic symptoms or an acute episode of hematemesis or melaena. It most commonly occurs in patients above 40 years of age. Telangiectasia can occur throughout the gastrointestinal tract. However, it is more common in the stomach or duodenum than in the colon. They are almost similar to mucocutaneous telangiectasia in size and appearance, and may be surrounded by an anaemic halo. Less commonly, AVMs and aneurysms occur. Our patient's capsular endoscopy exhibited a telangiectatic spot in the small intestine without bleeding.

Clinically important AVMs can occur in a number of organs, such as the lung, brain, and liver. While single AVMs can occur sporadically in the normal population as well as in patients with HHT, the presence of multiple AVMs in an organ such as the lung or brain make a sporadic aetiology less likely.

Additionally, CT-based research screening programmes of asymptomatic individuals have identified pancreatic AVMs at surprisingly high frequencies.(5)

Pulmonary arteriovenous malformations (PAVMs) provide a direct capillary-free communication between the pulmonary and systemic circulations. Patients with PAVMs are at risk of complications, most commonly cerebrovascular events due to embolic phenomena, with embolic material invading the filtering function of the pulmonary capillaries and reaching the central nervous system. Due to these right-to-left shunts, pulmonary arterial blood cannot be oxygenated, causing hypoxemia. Hypoxemia results in secondary polycythemia.(6) Catastrophic embolic cerebral events (embolic stroke, transient ischemic attack, and brain abscess) occur in patients with clinically silent PAVMs necessitating the need for early diagnosis and interventions such as embolization of PAVMs and antibiotic prophylaxis for some interventional procedures, especially dental.(7)

Although splenic involvement has been described in HHT, splenic abscess as a complication of HHT is seldom reported. However, an isolated PAVM causing a splenic abscess along with a splenic infarction has been described. Splenic infarcts and abscesses probably occur through the same mechanism of paradoxical embolism. Hence, this case is a rare manifestation where a splenic abscess has occurred in HHT.(9)

The diagnosis of HHT is made clinically on the basis of the Curaçao criteria. The four clinical diagnostic criteria are as follows: Epistaxis, telangiectasias, visceral lesions, arteriovenous malformations (AVMs), and a first-degree relative with HHT. These criteria define HHT as "definite" (three or four criteria), "suspected" (two criteria), and "unlikely" (zero or one criterion). The diagnosis may be established or confirmed by finding a pathogenic mutation in ENG, ACVRL1, SMAD4, or GDF2 genes.(8) Our patient fulfilled this criteria to diagnose HHT.

Conclusion

HHT must be considered in a patient with recurrent nose bleeds and iron deficiency anaemia. It can affect multiple organs and cause life threatening bleeding manifestations which warrants inward treatment. Splenic abscess and hematoma can occur as complications of HHT which warrant urgent intervention.

Declarations

Author contributions

Dr Dikshaladevi Pathmanathan initiated this case report along with Dr Nisanthan Selvaratnam as the first contact medical officer under supervision of Dr Nalayini Jegetheesan and Dr Thambipillai Peranantharajah who helped in arranging necessary investigations and guided the management.

Conflicts of interest

The authors declare that they have no conflicts of interest to be addressed regarding this case report.

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