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Case Report

Pulmonary arteriovenous malformation in a pediatric patient with epistaxis and hypoxemia

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ABSTRACT

Hereditary hemorrhagic telangiectasia (HHT; also known as Osler-Weber-Rendu syndrome) is an inherited vascular disorder with a spectrum of clinical manifestations depending on lesion distribution. Epistaxis, mucocutaneous telangiectasia, and gastrointestinal bleeding are most common. Patients with pulmonary arteriovenous malformations are at serious risk of cerebral embolic stroke and abscess due to paradoxical embolism, indicating the need for early diagnosis and intervention. Herein, we report a 14-year-old boy who presented to his pediatrician's office with hypoxemia and personal history of epistaxis, family history of HHT, and radiologic workup demonstrating pulmonary and cerebral arteriovenous malformations. He was diagnosed with HHT and treated by endovascular embolization.

Introduction

Hypoxemia is a condition of abnormally low levels of oxygen in the blood, which can have multiple causes, including right-to-left shunt, ventilation/perfusion (V/Q) mismatch, hypoventilation, diffusion limitations, and reduced inspired oxygen tension [1]. Right-to-left shunt occurs when blood passes from the right to left side of the heart without being oxygenated, and can be anatomic as in intracardiac shunts or pulmonary arteriovenous malformations (AVMs), or physiologic when perfused alveoli are not ventilated.

In patients with hypoxemia and a personal history of epistaxis or hemoptysis, family history of hereditary hemorrhagic telangiectasia (HHT), or exam findings revealing clubbing and/or cyanosis, there should be strong suspicion for a pulmonary AVM [2]. In patients with HHT, large AVMs can occur in the lungs (40%-60% of patients), liver (40%-70%), and brain/spine (10%) [3,4]. Radiologic workup for pulmonary AVM includes an initial chest x-ray, often followed by computed tomography of the chest, with contrast enhanced studies demonstrating enhancement of the feeding artery, aneurysm, and early-draining vein. Due to the potentially fatal complications of stroke and brain abscess due to paradoxical embolism, treatment of pulmonary AVMs with endovascular embolization is recommended as a safe and effective procedure for reducing morbidity and mortality in these patients [5,6].

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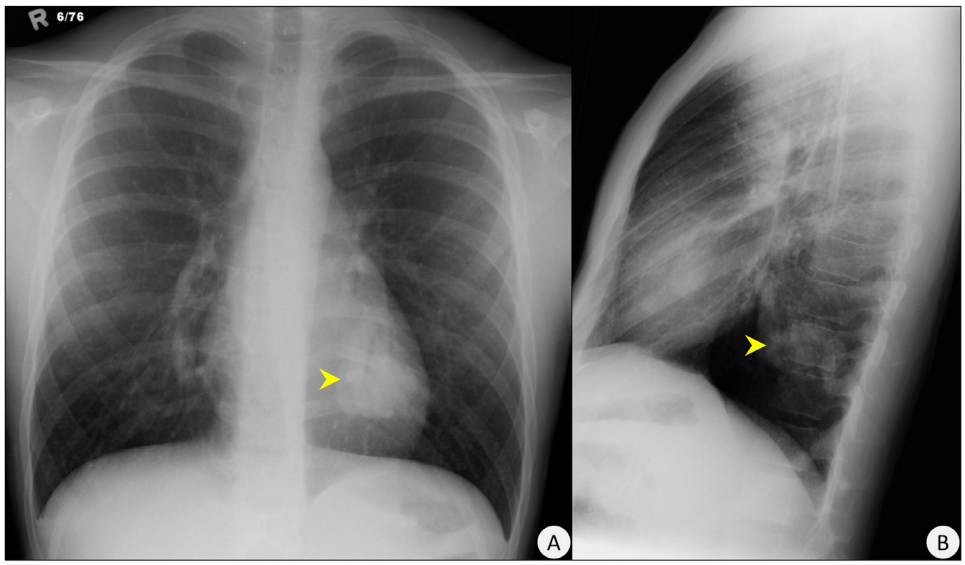

Fig. 1 – Posterior-anterior (A) and lateral (B) chest x-ray demonstrated retrocardiac mass suspicious for vascular malformation.

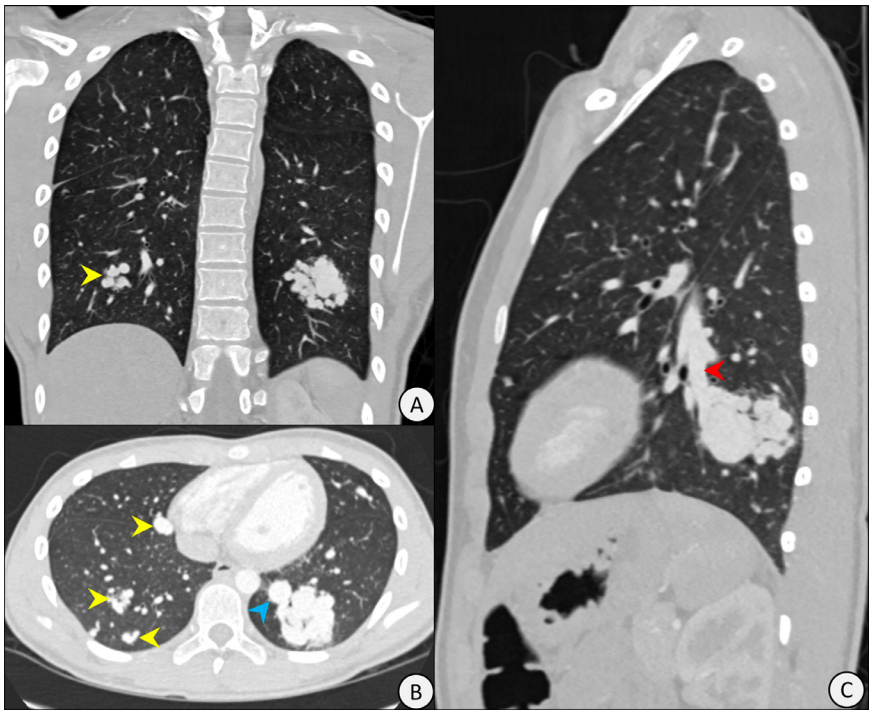

Fig. 2 – Computed tomography of the chest revealed multiple pulmonary arteriovenous malformations (AVM) (2A and 2B, yellow arrowheads), including large left lower lobe AVM with large arterial feeding vessel (2C, red arrowhead) and early-filling pulmonary vein (2B, blue arrowhead). (Color version available online.)

Case report

A 14-year-old male with history of mild epistaxis presented to his pediatrician's office for a routine well-visit where he was found to have blue nails and hypoxemia with oxygen saturation of 75%. He was referred to the emergency department for further evaluation, with mild improvement of his hypoxemia to 83% on 3L oxygen. Physical exam was notable for moderate finger clubbing. Family history was notable for a paternal uncle with HHT. Initial laboratory tests were normal. Chest x-ray demonstrated a 6 × 5 cm retrocardiac mass that was concerning for a vascular malformation (Fig. 1). Follow up CT chest showed multiple pulmonary AVM in both lungs, the largest

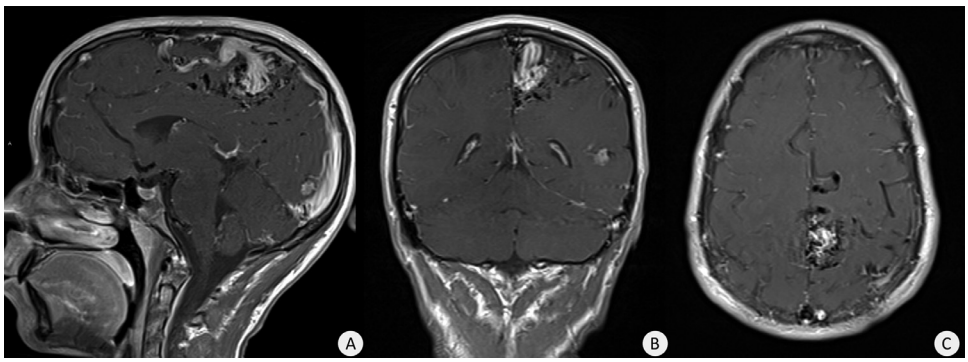

Fig. 3 – T1 contrast enhanced magnetic resonance imaging of the brain (3A-C) showed multiple cerebral arteriovenous malformations (AVM), the largest measuring 3.5 cm in the medial left parietal lobe supplied by both anterior and middle cerebral arteries, with predominantly superficial venous drainage.

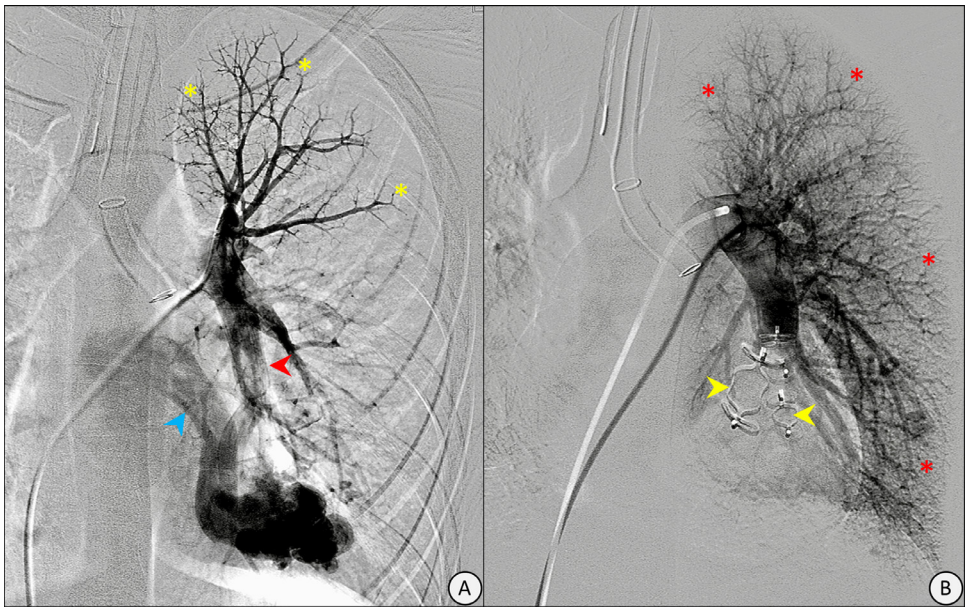

Fig. 4 – Pulmonary angiography (4A) demonstrated a large complex arteriovenous malformation (AVM) in the left lower lobe with large feeding vessels (red arrowhead) early pulmonary venous filling (blue arrowhead), and decreased perfusion to the lung parenchyma as shown by rapid tapering of peripheral pulmonary vessels (yellow asterisks). Repeat pulmonary angiography (4B) following successful embolization of the pulmonary AVM with Amplatzer plugs (yellow arrowheads) shows increased perfusion to the surrounding lung parenchyma (red asterisks). Oxygen saturation increased from 70% preprocedure to 92% postprocedure. (Color version available online.)

in the left lower lobe with feeding arteries measuring up to 9 mm in diameter (Fig. 2). Additional imaging workup with MRI brain revealed numerous cerebral AVMs, the largest measuring 3.5 cm in the medial left parietal lobe supplied by both anterior and middle cerebral arteries, with predominantly superficial venous drainage (Fig. 3). A clinical diagnosis of HHT was made. Genetic testing returned an ENG gene mutation, consistent with this diagnosis.

Four days following initial presentation, the patient was seen by interventional radiology for pulmonary angiography, which demonstrated a large complex AVM in the left lower lobe with large feeding vessels, early pulmonary venous filling, and decreased perfusion to the lung parenchyma as shown by rapid tapering of peripheral pulmonary vessels (Fig. 4, A). Successful embolization of the AVM was performed using Amplatzer plugs, and postembolization angiography revealed markedly increased lung perfusion (Fig. 4, B). Oxygen saturation increased from 70% preprocedure to 92% postprocedure.

At 2-year follow up, the patient had undergone 10 subsequent embolizations for pulmonary AVMs. Chest x-ray demonstrated resolution of retrocardiac mass with numerous embolization plugs and coils in the lower lungs (Fig. 5), oxygen saturation was 97%, and he denied dyspnea on exertion.

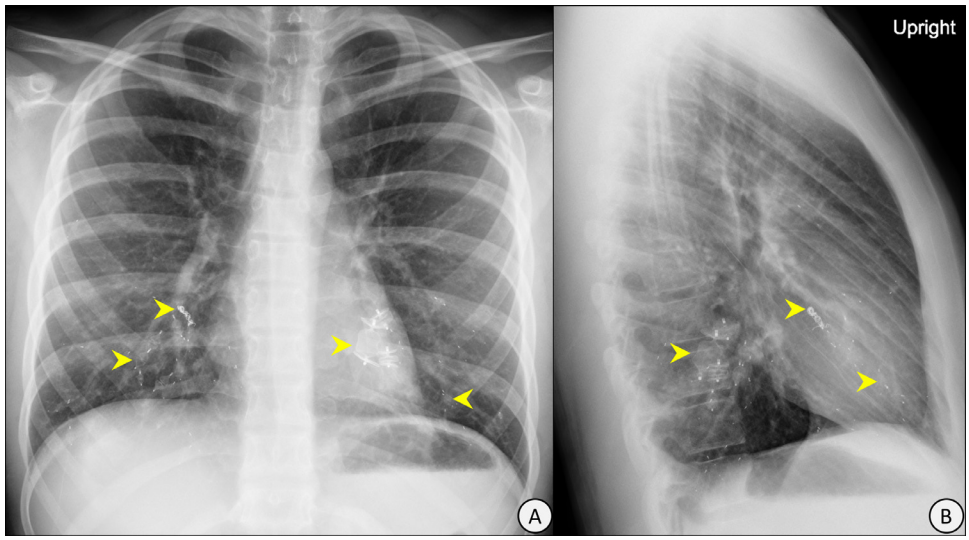

Fig. 5 – Two-year follow up posterior-anterior (A) and lateral (B) chest x-ray reveals numerous embolic coils and plugs (yellow arrowheads), with resolution of the retrocardiac mass.

Discussion

HHT (also known as Osler-Weber-Rendu syndrome) is an inherited vascular disorder with a spectrum of clinical manifestations depending on lesion distribution. Epistaxis, mucocutaneous telangiectasia, and gastrointestinal bleeding are most common. While 5 gene mutations have been identified as causing HHT, the vast majority (98%) are caused by mutations in the ENG (endoglin) and ACVRL1 (activin A receptor type II-like 1 gene), which lead to types 1 and 2 HHT, respectively [7]. Clinical diagnosis of HHT is based on the Curaçao diagnostic criteria, which includes a history of recurrent epistaxis, multiple telangiectasias, involvement of visceral lesions, and a first-degree relative with HHT, where the diagnosis is considered "definite" if 3 or more criteria are present, and "suspected" if 2 are present [6].

A multidisciplinary approach is needed to evaluate the many potential manifestations of HHT in order to achieve a complete diagnostic picture and therapeutic plan. Mortality in these patients increases with involvement of visceral organs, including AVM in the lung, brain, liver, and gastrointestinal tract [8]. Patients with pulmonary AVM in particular, as in this patient, are at serious risk of cerebral embolic stroke and abscess due to paradoxical embolism, indicating the need for early diagnosis and intervention [9].

Author's contributions

All authors were involved in the care of the patient, and the writing and editing the manuscript. Written consent for publication was obtained from the patient and parents.

Conflicts of interest

None.

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No funding was used for this study. No author was paid to write this article. Authors had full access to all data in the study.

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