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Case Report

The role of nuclear medicine in a case of Rendu–Osler–Weber disease with pulmonary involvement

ABSTRACT

Rendu–Osler–Weber syndrome or hereditary hemorrhagic telangiectasia (HHT) is a rare systemic disease. Its primary pathogenic expression is multiple arteriovenous malformations (AVM) and severe hypoxia. A case of suspected pulmonary embolism in a 49-year-old male with intestinal, cardiac, and pulmonary HHT affection is reported. Pulmonary AVM could create an apparent mismatch perfusion defect evident upon ventilation and perfusion scan (V/Q scan), leading to misinterpretation. It reinforces the importance between clinics, anatomy, and functional evaluation. Care must be taken when interpreting V/Q scan and the reporting physician must be alert to the possible sources of errors.

Keywords: Arteriovenous malformations, hereditary hemorrhagic telangiectasia, Rendu–Osler–Weber disease, technetium Tc 99 m aggregated albumin, ventilation-perfusion scan

INTRODUCTION

Rendu–Osler–Weber syndrome or hereditary hemorrhagic telangiectasia (HHT) is a rare systemic disease, affecting approximately 0.02% of the population.[1] Its primary pathogenic expression is multiple arteriovenous malformations (AVM), especially in lungs, liver, brain, and rarely the spine.[2] The incidence is 1:5000–1:10000 worldwide.[2] Common manifestations are telangiectases, reported in 74% of patients,[3] in nasal mucosa, gastrointestinal tract, lips, and hands. Over 90% of all HHT patients present chronic epistaxis due to nasal telangiectases by the age of 45 years.[4] Chronic bleeding from gastrointestinal telangiectases develops in at least 20% of patients.[4] Pulmonary AVM (PAVM) and hepatic AVM have been detected in 24%–40% and 41%–84% of HHT patients, respectively.[5]

CASE REPORT

A 49-year-old male, presenting at the emergency room with weakness, adynamia, fatigue evolving with hypoxemia, and progressive need for oxygen therapy. The patient reported recurrent epistaxis in childhood, hemorrhoidal disease, and refractory to iron replacement anemia. Transthoracic echocardiography showed right chambers significant dilation and pulmonary hypertension (pulmonary artery pressure: 71 mmHg). Due to pulmonary embolism suspicion, V/Q scan was performed. The ventilation study was carried out after ^{99m}Tc-labeled aerosols (^{99m}Tc-FITATE) and showed homogeneous distribution of the material in both the lungs. For perfusion scintigraphy, intravenously injected macroaggregate of ^{99m}Tc-labeled human albumin (^{99m}Tc-MAA)

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was used. Perfusion scan revealed moderately heterogeneous distribution of the material in both lungs, due to multiple radiopharmaceutical retention points, associated with filling failure areas in pulmonary periphery (mismatch altered areas with normal ventilation study). Due to possible ^{99m}Tc-MAA particles aggregation resulting in artifacts because of blood withdrawal into the syringe, new perfusion scan was carried out 48 h later and the same image pattern was observed. [Figure 1]. Meanwhile, computed tomography angiography of the pulmonary artery (CTPA) ruled out pulmonary thromboembolism (PTE), demonstrating multiple aneurysms in pulmonary artery segmental branches [Figure 2]. The clinical scenario of the past epistaxis associated with visceral AVM substantiated the proposed diagnosis of HHT. Hotspots persistence in pulmonary perfusion scan were interpreted as areas of ^{99m}Tc-MAA retention in arterial segments branches aneurysms and mismatch altered peripheral areas were interpreted as pitfalls caused by congenital lung vascular anomalies secondary to the underlying disease. Currently, the patient is being followed up in an outpatient clinic for pulmonary hypertension cases.

DISCUSSION

V/Q scan is considered one of the two mainly used imaging modalities for the diagnoses of PTE.[6] Ventilation scintigraphy can be evaluated by ^{99m}Tc-labeled aerosols and perfusion by intravenously injected macroaggregates of ^{99m}Tc-MAA, which diameter of 15–100 μm leads to microembolization of pulmonary precapillary arterioles and capillaries.[6] V/Q mismatch defects of at least one segment or two subsegments wedge-shaped with the base projecting to the lung periphery is criteria for PTE presence. V/Q scan needs to be interpreted in the context of the clinical likelihood for PTE.[6,7] The occurrence of a false positive scan is an unusual event, but it is vital that the nuclear physician has the knowledge of a number of sources of errors when reporting the study.[6,7] Congenital lung vascular anomalies is described as one of the causes of segmental mismatches, consequently a potential pitfall in the interpretation of V/Q scan.[6] Single photon-emission computed tomography is the mainly used nuclear imaging modality to rule out PTE.[6]

PAVM are the abnormal connections between pulmonary arteries and pulmonary veins. Most patients with PAVM have the autosomal dominant HHT disease. HHT is recognized through Curaçao criteria, a triad of cutaneous telangiectasia, family history of the disorder, and recurrent epistaxis.[8] The fundamental defect comes from right-to-left shunting. Deoxygenated blood from the pulmonary artery is shunted into the pulmonary vein, which carries oxygenated blood into the left atrium. If the right to left shunt is >20% of the systemic cardiac output, the patient can then develop cyanosis, clubbing, and polycythemia.[8,9]

Imaging exams are performed according to the patient's clinical presentation and may include abdominal ultrasonography, plain radiography, and eventually, chest computed tomography.[10] In light of the above, the most frequent situations related to this rare syndrome in the radiologist's daily routine include incidental finding of visceral AVM in patients without previous diagnosis of HHT.[10] In the present case, vascular pulmonary lesion was incidentally found on CTPA, described as arteriovenous fistulas in bilateral basal branches.

HHT with pulmonary AVM should be considered in patients with severe hypoxia due to right-to-left shunting and the presence of multiple telangiectases.[7]

This case illustrates the risk of misinterpreting a V/Q scan with unmatched perfusion defects in a patient with hemorrhagic telangiectasia.

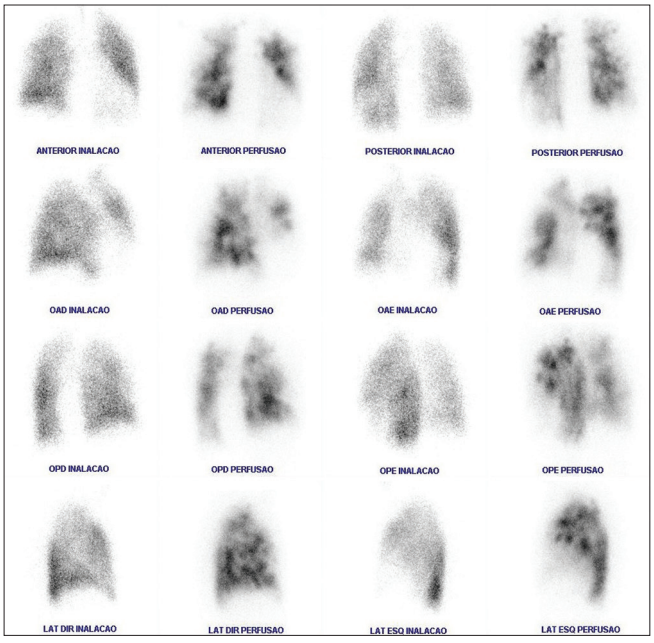

Figure 1: Ventilation-perfusion scintigraphy

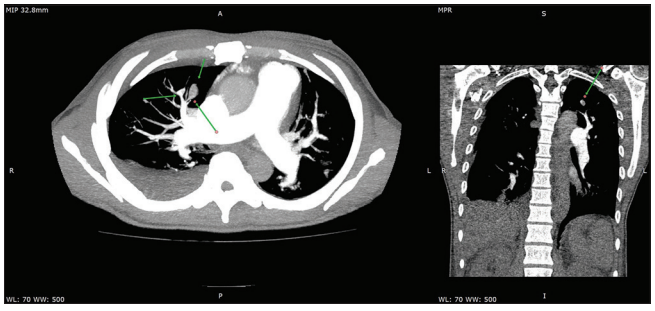

Figure 2: CTPA showing aneurysms in pulmonary artery segmental branches

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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