Hereditary hemorrhagic telangiectasia (HHT): Whole body imaging findings with emphasis in central nervous system involvement

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Learning objectives

1) To illustrate the spectrum of imaging abnormalities in HHT.

2) To understand the pathogenesis of development of central nervous system lesions.

3) To determine the current role of Computed Tomography, Magnetic Resonance Imaging and Digital Subtraction Angiography (DSA) in their detection, characterization and treatment.

Background

HHT or Rendu-Osler-Weber (ROW) disease is an autosomal dominant disorder with variable penetrance caused by mutations of endoglin or activin receptor-like kinase, involved in the angiogenic process.

In patients with ROW disease there is a lack or a dysfunction of the capillaries, so that arteries connect directly into veins, creating weakened bridges that can easily rupture and bleed, or a shunting which may cause other complications depending on their location. The two types of abnormal blood vessels in ROW are telangiectases (small size blood vessels) and arteriovenous malformations (AVMs) involving larger vessels.

ROW disease is characterized by epistaxis, mucocutaneous telangiectases (Fig. 1) and visceral AVMs. Although in this disease epistaxis and other mucocutaneous telangiectases are the most common clinical manifestations, visceral AVMs can lead to the life threatening complications.

Prevalence is estimated to be approximately 1:10.000.

Diagnostic criteria for a definitive diagnosis include at least three of the following four:

1. Spontaneous and recurrent epistaxis.

2. Multiple mucocutaneous telangiectases.

3. Visceral lesions such as gastrointestinal or pulmonary AVM.

4. A first-degree relative with ROW disease.

Images for this section:
Fig. 1: Multiple tongue and lip telangiectases in a patient with ROW disease (a). Selective external carotid DSA of another patient showing diffuse telangiectases (b).
Imaging findings OR Procedure details

The most commonly affected organs are the lungs, brain, liver and gastrointestinal tract, although virtually any body system can be affected.

**LUNG**

Lung manifestations in ROW disease include AVMs and telangiectases. Pulmonary AVM is the most frequent lesion in these patients and are usually found in up to 25%-50%. Most are asymptomatic and neurologic complications due to paradoxical embolism are often the initial clinical presentation.

At standard chest radiograph pulmonary AVMs appear as well defined nodular lesions, located in the central or peripheral lung with linear stria connecting to the ipsilateral pulmonary hilium (Fig. 2). This linear stria represents the tortuous feeding arteries and draining veins. However chest radiograph has a low sensitivity because many pulmonary AVMs are too small to be detected. The most frequent involvement of the basal and posterior segments of the lower lobes is another cause of underdiagnosis.

Currently unenhanced helical CT scan with thin collimation and maximum intensity projection (MIP) reconstruction is the technique of choice in detection of pulmonary AVM in patients with ROW disease (Fig. 3). There are two types of pulmonary AVM: the simple type (90%), consisting of one feeding artery into an aneurysm sac with one draining vein, and the complex type (10%), in which there are multiple feeding and draining vessels.

Multidetector thoracic CT has replaced DSA as the reference imaging method. DSA is mainly used to endovascular treatment of pulmonary AVMs (coiling). Intraarterial embolization is generally indicated for lesions where feeding arteries are larger than 3mm, even when symptom free, due to potential life-threatening complications (Fig. 4).

**BRAIN AND SPINE**

Neurologic manifestations appear in about 10% of patients with ROW disease but rarely are the initial manifestation of the disease. Neurological clinical manifestations are more often a consequence of pulmonary AVM with paradoxical embolism (ischemia, infections) and hepatic AVM with porto-systemic shunt (hepatic encephalopathy), than directly related to a complication of brain or spinal vascular malformations.

In these patients pulmonary AVM is well recognized as a cause of paradoxical brain embolism. Emboli that circulate through an AVM may bypass the pulmonary capillary bed and be carried to the brain or to the spinal cord, resulting in ischemia or infection secondary to thrombus and septic emboli respectively.

Ischemia is the most frequent neurologic manifestation, typically in different topographies and timings.

Infection is the most serious neurologic manifestation. It is estimated that 5% of patient with pulmonary AVM develop a brain abscess, usually lodged in the cortex at the rich capillary plexus of the gray-white matter junction. Moreover in these patients hypoxemia decreases resistance of cerebral tissue to infection.
and abscesses are typically multiple and recurrent (Fig. 5). Exceptionally abscesses may occur in the spinal cord (Fig. 6).

Complete eradication of the pulmonary AVM is essential because the neurologic complications will recur unless this is achieved.

Hepatic encephalopathy due to intrahepatic porto-systemic shunts is another potential complication in patients with ROW disease. This metabolic disorder appears in magnetic resonance imaging as a hyperintensity in the basal nuclei on T1-weighted images (Fig. 7).

Only one third of central nervous system findings are in relation to central nervous system vascular malformations (brain and spinal cord) (Fig. 8). Magnetic resonance imaging is usually the first radiological approach but cerebral DSA may be required for diagnosis of equivocal lesions and specially for planning the treatment (surgery, embolisation and/or stereotactic radiosurgery).

**LIVER AND GASTROINTESTINAL TRACT**

Abdominal findings in ROW disease are predominantly within the liver. Imaging can be performed using a variety of techniques (sonography, MRI, CT, DSA). However, multidetector CT is probably the best.

Most of liver involvement is asymptomatic: telangiectasia and hepatic perfusion abnormalities. They are frequently diffuse and are best seen during the arterial phase with multidetector CT and MRI. Biliary abnormalities such as cysts, strictures and dilatations are also reported. Telangiectasia is the most commonly seen lesion, appearing as focal hypervascular lesions of variable size, rounded or resembling a stellate mass. If numerous, the liver appears diffusely heterogeneous (Fig. 9). Often the small lesions are best displayed on CT coronal MIP images obtained in early arterial phase.

Symptomatic liver involvement is less frequent. Lesions of HHT may cause bleeding or result in shunting. Angiodysplastic lesions of HHT can result in direct communication between arteries and veins. Shunts from the hepatic artery to the hepatic veins, portal veins or both can produce high-output heart failure or portal hypertension. Arteriovenous shunts are detected during the arterial phase on dynamic CT or MRI when early opacification of the hepatic veins is observed. Images can show both hepatic arteries and hepatic veins enlarged. Portovenous shunts are better seen in the hepatic phase (dilated portal vein communicating with the hepatic vein). Sonographic doppler findings include abnormal flows of vessels affected, abnormal echogenicity of the liver parenchyma and dilatation of the arteries and hepatic veins (Fig. 10).

Presence of telangiectases and dilatation of the common hepatic artery can be considered pathognomonic for HHT in the presence of a compatible clinical history.

Other vascular dysplasic lesions may occur in any abdominal organ, with pancreatic AMVs being the most common.

**Images for this section:**
Fig. 1: Posteroanterior (a) and lateral (b) chest radiograph show a well defined nodule in the right lower lobe with feeding and draining vessels (arrows). Note increased density of the right hilium.

Fig. 2: Axial (a) and coronal (b) MIP reconstruction of a chest CT scan show two right lower lobe pulmonary AVMs in a patient with ROW disease.
Fig. 3: DSA during intrarterial embolization of a simple pulmonary AVM (a and b). Anteroposterior chest radiograph after embolotherapy shows coils occluding two pulmonary AVMs in the right lung (c).
Fig. 4: Cervical spinal cord MRI sagittal T2wi (a) and T1wi after gadolinium injection (b) in a patient with ROW disease and pulmonary AVMs show an intramedullar abscess at C5-C7 level. Note rim enhancement (arrow in b).
**Fig. 5:** Coronal T1-weighted brain MRI with bilateral hyperintensity in the basal nuclei in a patient who presented behavior changes (a). Selective splenic DSA in portal phase showed multiple porto-systemic shunts (b).

**Fig. 6:** CT scan after intravenous contrast injection in the same patient as figure 4, shows a space-occupying lesion with vasogenic edema and ring enhancement in right parietal lobe (a). Two weeks later in another scan appeared a new abscess in the same lobe (b).
**Fig. 7:** Figure 8a. DSA at early arterial phase showing angiomatous blush (arrow) of the distal right PICA. Note the absence of draining vein and other signs of arteriovenous shunt which suggests the diagnosis of telangiectasia. Figure 8b. Selective left vertebral artery DSA, showing a perimedullar arteriovenous fistula in the spinal cord (red arrow pointing the pathologic blush and blue arrow at the early venous drainage).
Fig. 8: Heterogeneous enhancement of the liver parenchyma due to perfusion abnormalities (blue arrow) and telangiectases (red arrow).
Fig. 9: Hepatic ultrasound shows dyshomogeneous parenchyma and dilatation of the hepatic artery (red arrows). Colour and spectral doppler evaluation (c) shows an increased and turbulent flow in the hepatic artery.
Conclusion

Although HHT is a rare disease and usually the most common clinical presentation are related with peripheral hemorrhage such as epistaxis or gastrointestinal bleeding, its imaging spectrum includes central nervous system lesions which are being recognized with increasing frequency.

Radiologist need to be familiar with its wide spectrum of general and neurologic findings which may lead to potentially life-threatening complications.

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References


