Introduction

Hereditary hemorrhagic telangiectasia (HHT) is a genetic disorder that causes vascular abnormalities throughout several organ systems. The diagnosis is clinical and often missed -- it has been estimated that only about 10% of all HHT patients have been formally diagnosed. Vascular malformations can be found in any organ system, but most commonly in central nervous system, pulmonary vasculature, hepatic vasculature, and mucous membranes.

Case Presentation

- 47 year old woman with a medical history of iron deficiency anemia presented to pulmonary clinic in consultation regarding a pulmonary arteriovenous malformation (pAVM).
- pAVM had been diagnosed several years prior after vehicular accident after which she had significant hemoptysis and required endovascular coiling.
- Follow up CTA showed 1.6x1.5 cm aneurysm of right middle lobe, 5mm aneurysm of right lower lobe, 2 other AVMs of right upper and lower lobes without aneurysm formation (Figure 1, as a representation of one such AVM).
- On review of systems, she endorsed recurrent epistaxis of both nostrils since childhood, heavy menstrual bleeding, and dyspnea on exertion.
- Physical exam revealed small vascular-appearing lesions on her tongue, lips, and chest.
- She noted that her daughter had similar appearing lesions around her mouth.
- Given the presence of 3 of 4 of the Curacao criteria, the patient was given a definite diagnosis of HHT.

Discussion

- Diagnosis of HHT is clinical and based on the Curacao criteria, the four components of which include spontaneous or recurrent epistaxis, multiple characteristic MCTs, visceral AVMs, and a diagnosed first-degree relative.
- Number of criteria met <2 infers unlikely diagnosis, 2 infers possible diagnosis, and ≥3 infers definite diagnosis of HHT.
- Once diagnosis is made, all patients should be screened for clinically significant AVMs.
- Patients should have full ENT evaluation, especially with recurrent or spontaneous epistaxis.
- Endoscopic evaluation of gastrointestinal telangiectasias and AVMs should be performed if there is significant anemia not accounted for by degree of epistaxis.
- Central nervous system AVMs should be evaluated by MRI and MRA of the brain; patients with AVMs >1cm in diameter should have neurosurgical evaluation for potential embolization and/or radiosurgery.
- Transthoracic echocardiogram with bubble study, followed by CT-chest if appropriate, is used to evaluate for cardiopulmonary abnormalities and shunting; if present, then antibiotic prophylaxis before dental and surgical procedures is required, given significantly higher risk of intracranial infection.
- Abdominal ultrasound is used to screen for hepatic AVMs, and if present, should undergo CT or MRI of the abdomen for further characterization.

Conclusion

Though HHT is a relatively rare diagnosis, it confers significant morbidity and mortality, especially when undiagnosed. Providers should have a low threshold for further investigation when characteristic findings are seen in patients or family members, as the vascular lesions can have a significant clinical impact.

References