Arteriovenous Malformations in Osler Weber-Rendu: A Patient Study

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Objectives

- Learn what an arteriovenous malformation (AVM) is.
- Learn imaging modalities that can be used to image AVM.
- Learn the typical appearance of AVM.
- Appreciate the clinical manifestations of AVM in Osler-Weber-Rendu/HHT.
Mrs. R: History

• 37 y/o woman w/ R. MCA ischemic stroke

• Coagulation studies were negative/normal

• No ASD/PFO (potential cause of paradoxical embolism)

• “Cryptogenic stroke” presentation (no known cause)
Mrs. R: AP CXR

FINDINGS

- Rounded opacity at R. cardiophrenic angle
- Linear opacity extending between rounded opacity and hilum

Findings suggestive of PAVM
General Architecture of Arteriovenous Malformations (AVM)

- Dilated, tortuous feeding and draining vessels
- A “nidus” or “serpiginous” mass of malformed vessels
- Diminished oxygen diffusion (A-V shunt); abnormal/absent capillary bed
- Rapid filling and emptying

http://neuro.wehealny.org
Imaging Pulmonary AVM: Menu of Tests

- Chest X-ray
  - Only useful for large AVM
- Chest CT (+/- C)
  - Gold standard
- Pulmonary angiography
  - Used mainly for therapeutic interventions due to invasiveness
- MR angiography
- Transthoracic Contrast Echo
  - Shunt quantification
Features of Pulmonary AVM on Chest X-Ray

- Round or oval opacity
- Uniform density, noncalcified
- Often lobulated but sharply defined
- **Location:** frequently in lower lobes
- **Connection:** may show feeding artery radiating from hilus
Features of Pulmonary AVM on CT

• Homogeneous, non-calcified, circumscribed nodule or serpiginous mass
• MUST have a direct feeding artery
  – Dilated efferent draining vein
• With contrast:
  – Sequential enhancement of feeding artery and aneurysmal part
  – Early filling of draining vein (to differentiate from highly vascular soft tissue mass)
Mrs. R: Chest CT

- Feeding branch of pulmonary artery
- R. hemidiaphragm
- Nidus of AVM
Mrs. R: Decision to Embolize

With the clear determination of pulmonary AVM on CT, it was decided to proceed with therapeutic embolization.
Mrs. R: Digital subtraction pulmonary arteriogram

(L. Lung, normal)
Mrs. R: Digital subtraction pulmonary arteriogram

(R. Lung, before therapeutic embolism)
Therapeutic Embolism of PAVM

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Mrs. R: Post-embolization

Digital subtraction pulmonary arteriogram (R. Lung, post-therapeutic embolism)
Mrs. R: Post-embolization

Digital subtraction pulmonary arteriogram (R. Lung, post-therapeutic embolism)

Pre-embolism:
- R. pulmonary artery
- AVM nidus

Post-embolism:
- No filling in embolized AVM
Differential Diagnosis of Pulmonary AVM

Congenital (~90%)

- Hereditary Hemorrhagic Telangiectasia (Osler-Weber-Rendu)
  - Type I – EDG1 mutations
  - Type II – ALK1 mutations
  - Other – unmapped loci

Acquired

- Highly vascular parenchymal lesion (compare kinetics with other vascular structure)
Clinical Manifestations of Pulmonary AVM

Primary mechanism

- Dyspnea on exertion
- Hypoxemia

right-left shunt

- Stroke
- Transient ischemic attack
- Cerebral abscess

paradoxical embolism (our patient)

Most PAVM are asymptomatic for years, so many are first encountered incidentally on imaging
Screening for Pulmonary AVM in HHT Patients

Chest Radiographs
- 

Contrast echocardiography
- + 

Absence of PAVM

Absence of PAVM

Positive PAVM

Prophylactic antibiotics to prevent brain abscess from paradoxical emboli
Hepatic AVM: Menu of Tests

- CT
  - Specific, gold standard
- Doppler Color Ultrasound (Echo Color Doppler)
  - Low cost
  - No radiation
  - Highly sensitive
Hepatic AVM on CT

- Early Arterial Phase
- Late Arterial Phase
- Portal Venous Phase

→ AVM

△ hepatic veins

• AVM is most apparent in early arterial phase, no longer visible in portal venous phase
• Hepatic veins fill early

Hepatic AVM Features on CT

- **Heterogenous opacification of liver**
  - Due to many small hypervascular spots (in HHT patients)

- **Early arterial phase contrast filling of AVM**

- **Hepatic vein enhancement in early arterial phase**
  - Caused by AVM shunt
Hepatic AVM Features on US

- **Hypervascularization: tortuous hepatic arteries**
  - Highly visible main intrahepatic branches of the hepatic artery in the portal spaces.
  - Shunt determines tortuosity and enlargement of these vessels.


EPA=hepatic artery  
PV=portal vein
Hepatic AVM Features on US

- **Intrahepatic “color spots”**
  - The presence of subcapsular vascular spots near the hepatic capsule consisting mainly of arterial blood flow.

Osler-Weber-Rendu/Hereditary Hemorrhagic Telangiectasia (HHT)

• One of the most common monogenic diseases (~1 in 5000-10000)

• Progressive clinical manifestations over decades

• Epistaxis
  – Usually 1st manifestation, ~10 yr of age

• Telangiectasia
  – Onset at ~3rd decade of life
  – Lips, tongue, palate, fingers, and face
Diagnosis of HHT

• Curacao criteria
• Definite diagnosis with 3 out of 4 criteria

<table>
<thead>
<tr>
<th>Criterium</th>
<th>Description</th>
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<tbody>
<tr>
<td>Epistaxis</td>
<td>Spontaneous, recurrent nose bleeds; often first manifestation</td>
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<tr>
<td>Telangiectasias</td>
<td>Multiple, at characteristic sites: lips, oral cavity, fingers, nose</td>
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<tr>
<td>Visceral lesions</td>
<td>Gastrointestinal telangiectasias (+/- bleeding)</td>
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<tr>
<td></td>
<td>Pulmonary AVM</td>
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<td>Hepatic AVM</td>
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<td>Cerebral AVM</td>
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<td>Spinal AVM</td>
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<tr>
<td>Family history</td>
<td>A first-degree relative with HHT according to these criteria</td>
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</tbody>
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Gossage et al. *Am J Respir Crit Care Med* 1998; 158: 643-661
Likely locations of AVM

- lung
- liver
- brain
- spinal cord

Osler-Weber-Rendu/HHT

- spleen
- kidney
- spermatic cord

Idiopathic/Other
Imaging AVM: Summary

1. Menu of Tests to Image AVM is Organ-specific
   • Pulmonary: CT, Angiography (for interventions)
   • Hepatic: CT, US

2. Typical architecture of AVM:
   • Entirely vascular structure that ‘shortcuts’ capillaries
   • Dilated, tortuous feeding and draining vessels
   • Rapid filling and draining of contrast

3. Detecting an AVM may provide an opportunity to pre-emptively address it and offer genetic counseling for Osler-Weber-Rendu patients and their family
References

Acknowledgements

• Salomao Paintuch, MD
• Dmitri J Rabkin, MD
• Gillian Lieberman, MD
• Larry Barbaras (webmaster)
• Maria Levantakis