Case Studies: Morphology of Aortic Regurgitation, Root Involvement

Vera H. Rigolin, MD, FASE, FACC, FAHA
Professor of Medicine, Northwestern University’s Feinberg School of Medicine
Medical Director, Echocardiography Laboratory
Northwestern Memorial Hospital
President, American Society of Echocardiography

No Disclosures
Introduction

• Aortic regurgitation may be caused by primary disease of the aortic valve leaflets and/or the wall of the aortic root.

• AR due to dilatation of the ascending aorta is now more common than primary valve disease in pts undergoing AVR for isolated AR.

Anatomic Location

1. Aortic sinuses of Valsalva
2. Sinotubular junction
3. Mid ascending aorta (midpoint in length between Nos. 2 and 4)
4. Proximal aortic arch (aorta at the origin of the innominate artery)
5. Mid aortic arch (between left common carotid and subclavian arteries)
6. Proximal descending thoracic aorta (begins at the isthmus, approximately 2 cm distal to left subclavian artery)
7. Mid descending aorta (midpoint in length between Nos. 6 and 8)
8. Aorta at diaphragm (2 cm above the celiac axis origin)
9. Abdominal aorta at the celiac axis origin

AHA/ACC 2010 Guidelines for Thoracic Aortic Disease
### Gene Defects Associated with Familial Thoracic Aortic Aneurysm and Dissection

<table>
<thead>
<tr>
<th>Defective Gene Leading to Familial Thoracic Aortic Aneurysms and Dissection</th>
<th>Contribution to Familial Thoracic Aortic Aneurysms and Dissection</th>
<th>Associated Clinical Features</th>
<th>Comments on Aortic Disease</th>
</tr>
</thead>
<tbody>
<tr>
<td>TGFB2 mutations</td>
<td>4%</td>
<td>Thin, translucent skin, Arterial or aortic tortuosity, Aneurysm of arteries</td>
<td>Multiple aortic dissections documented at aortic diameters &lt;5.0 cm</td>
</tr>
<tr>
<td>MYH11 mutations</td>
<td>1%</td>
<td>Patent ductus arteriosus</td>
<td>Patient with documented dissection at 4.5 cm</td>
</tr>
<tr>
<td>ACTA2 mutations</td>
<td>14%</td>
<td>Livedo reticularis, Iris fleckuli, Patent ductus arteriosus, Bicuspid aortic valve</td>
<td>Two of 13 patients with documented dissections &lt;5.0 cm</td>
</tr>
</tbody>
</table>

### Genetic syndromes associated with thoracic aortic aneurysm and dissection

<table>
<thead>
<tr>
<th>Genetic Syndrome</th>
<th>Common Clinical Features</th>
<th>Genetic Defect</th>
</tr>
</thead>
<tbody>
<tr>
<td>Marfan syndrome</td>
<td>Skeletal features (see text), Ectopia lentis, Dural ectasia</td>
<td>FBN1 mutations*</td>
</tr>
<tr>
<td>Loos-Dietz syndrome</td>
<td>Bifid uvula or cleft palate, Arterial tortuosity, Hypertelorism, Skeletal features similar to MFS, Craniosynostosis, Aneurysms and dissections of other arteries</td>
<td>TGFB2 or TGFB1 mutations</td>
</tr>
<tr>
<td>Ehlers-Danlos syndrome, vascular form</td>
<td>Thin, translucent skin, Gastrointestinal rupture, Rupture of the gravid uterus, Rupture of medium-sized to large arteries</td>
<td>COL3A1 mutations</td>
</tr>
<tr>
<td>Turner syndrome</td>
<td>Short stature, Primary amenorrhea, Bicuspid aortic valve, Aortic coarctation, Webbed neck, low-set ears, low hairline, broad chest</td>
<td>45,X karyotype</td>
</tr>
</tbody>
</table>
**Risk factors for thoracic aortic dissection**

**Conditions Associated With Increased Aortic Wall Stress**
- Hypertension, particularly if uncontrolled
- Pheochromocytoma
- Cocaine or other stimulant use
- Weight lifting or other Valsalva maneuver
- Trauma
- Deceleration or torsional injury (eg, motor vehicle crash, fall)
- Coarctation of the aorta

**Conditions Associated With Aortic Media Abnormalities**
- Genetic
  - Marfan syndrome
  - Ehlers-Danlos syndrome, vascular form
  - Bicuspid aortic valve (including prior aortic valve replacement)
  - Turner syndrome
  - Loeys-Dietz syndrome
  - Familial thoracic aortic aneurysm and dissection syndrome
- Inflammatory vasculitides
  - Takayasu arteritis
  - Giant cell arteritis
  - Behçet arteritis
- Other
  - Pregnancy
  - Polycystic kidney disease
  - Chronic corticosteroid or immunosuppression agent administration
  - Infections involving the aortic wall either from bacteremia or extension of adjacent infection

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**Mechanisms of AR due to abnormalities of the ascending aorta**

- Dilatation of the aortic annulus
  - separation of the leaflets
  - Tension and bowing of the leaflets result in thickening and retraction
- Aortic dissection
History

- 29 yr old female with Marfan’s syndrome
- Admitted with dyspnea and LE edema
- Diagnosed with an aortic aneurysm, chronic type A dissection and AR
- Presented to CT surgeon’s office with c/o chest pain
History

• PMH:
  – Multiple sclerosis
  – Asthma
  – Scoliosis
  – Dislocated lens

• FH
  – Mother with Marfan’s – died during aortic surgery
LVEDVI = 115 ml/m²
LVESVI = 58 ml/m²
LVEF = 50%
Pressure halftime = 239 msec

CT of the aorta

- Annulus: 2.5 x 3.3 cm
- Sinus of Valsalva: 5.3 x 5.2 x 5.3 cm
- Sino-tubular junction: 6 x 6 x 6.2 cm
- Mid ascending aorta: 6.7 x 6.4 cm
- Proximal aortic arch: 3.1 x 3.1 cm
- Distal aortic arch: 2.5 x 2.4 cm
- Lower descending thoracic aorta: 1.8 x 1.7 cm
Surgery

• Aortic root replacement with 27 mm Mechanical valve-graft
• Coronary reimplantation
• Resection and replacement of ascending aorta and total transverse arch using 24 mm dacron graft with reimplantation of inominate artery
• Hypothermic circulatory arrest with antegrade cerebral perfusion via right axillary artery cannulation
History

- 65 yr old Asian female who presents to the ER with chest pain
- Patient does not speak English and relays her complaint by pointing to her chest and moaning
- Patient initially thought to have ACS
- Echo ordered when murmur was heard
CT scan of the aorta

Stanford type A aortic dissection extending from the aortic valve to just beyond the origin of the left subclavian artery. The dissection also extends into the brachiocephalic artery.

Aneurysmal dilatation of the ascending aorta with a maximal dimension of 4.4 cm (non-orthogonal measurement).
Surgery

- Replacement of aortic root and ascending aorta with a 32 mm Gelweave graft
- Repair of the aortic valve

History

- 35 yr old male with a known heart valve problem since childhood
- He presented with a febrile illness 8 months prior. Blood cx positive for Strep
- Successfully treated with antibiotics
- Now feels well
MRA of the Aorta

There is aneurysmal dilatation of the mid ascending aorta.

The following orthogonal measurements of the thoracic aorta were obtained:
- Annulus: 3.2 x 3.3 cm
- Sinus of Valsalva: 4.1 x 4.8 x 4.0 cm. The largest dimension is between the left coronary cusp and the non-coronary cusp.
- Sino-tubular junction: 4.1 x 3.9 cm
- Mid ascending aorta: 5.0 x 5.1 cm
- Proximal aortic arch: 2.9 x 2.9 cm
- Distal aortic arch: 2.3 x 2.5 cm
- Lower descending thoracic aorta: 2.1 x 2.2 cm
Thank You