LVOTO AND AORTA IN CONGENITAL CARDIOVASCULAR DISEASES

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ESC Guidelines for the management of grown-up congenital heart disease (new version 2010)

The Task Force on the Management of Grown-up Congenital Heart Disease of the European Society of Cardiology (ESC)

Endorsed by the Association for European Paediatric Cardiology (AEPC)

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Left ventricular outflow tract obstruction (LVOTO)

- Valvular (75%)
- Subvalvular
- Supravalvular
- Multiple levels
Subvalvular stenosis (subAS)

- 6.5% congenital heart diseases
- Frequently associated with ASD, VSD and Shone’s complex
  (supravalve mitral membrane, parachute mitral valve, subaortic stenosis, aortic coarctation, bicuspid aortic valve)

- Fibrous ridge (membrane) – most frequent form of subAS (90% cases)
- Fibromuscular narrowing
- Additional mitral tissue (casuistic)
subAo membrane
Fibromuscular narrowing
Fibromuscular narrowing

\[ \Delta p = 4(v_2^2 - v_1^2) \]
### Table 10  Indications for intervention in subaortic stenosis

<table>
<thead>
<tr>
<th>Indications</th>
<th>Class</th>
<th>Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>Symptomatic patients (spontaneous or on exercise test) with a mean Doppler gradient $\geq 50$ mmHg or severe AR should undergo surgery</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td><strong>Asymptomatic patients should be considered for surgery when:</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>• LVEF is $&lt;50%$ (gradient may be $&lt;50$ mmHg due to low flow)</td>
<td>IIa</td>
<td>C</td>
</tr>
<tr>
<td>• AR is severe and LVESD $&gt;50$ mm (or $25$ mm/m$^2$ BSA) and/or EF $&lt;50%$</td>
<td>IIa</td>
<td>C</td>
</tr>
<tr>
<td>• mean Doppler gradient is $\geq 50$ mmHg and LVH marked</td>
<td>IIa</td>
<td>C</td>
</tr>
<tr>
<td>• mean Doppler gradient is $\geq 50$ mmHg and blood pressure response is abnormal on exercise testing</td>
<td>IIa</td>
<td>C</td>
</tr>
<tr>
<td><strong>Asymptomatic patients may be considered for surgery when:</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>• mean Doppler gradient is $\geq 50$ mmHg, LV normal, exercise testing normal, and surgical risk low</td>
<td>IIb</td>
<td>C</td>
</tr>
<tr>
<td>• progression of AR is documented and AR becomes more than mild (to prevent further progression)</td>
<td>IIb</td>
<td>C</td>
</tr>
</tbody>
</table>
Supravalvular aortic stenosis (supraAS)

- About 7% of LVOTO
- Frequently associated with Williams-Beuren syndrome (up to 80%)
- Hourglass deformity
- Localized fibrous diaphragm
- Diffuse hypoplasia of ascending aorta

„elfin” face
supraAS - TTE
Table 9  Indications for intervention in supravalvular aortic stenosis

<table>
<thead>
<tr>
<th>Indications</th>
<th>Class</th>
<th>Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patients with symptoms (spontaneous or on exercise test) and mean Doppler gradient ≥50 mmHg should undergo surgery</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>Patients with mean Doppler gradient &lt;50 mmHg should undergo surgery when they have:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• symptoms attributable to obstruction (exertional dyspnoea, angina, syncope) and/or</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>• LV systolic dysfunction (without other explanation)</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>• severe LVH, attributable to obstruction (not related to hypertension)</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>• when surgery for significant CAD is required</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>Patients with mean Doppler gradient ≥50 mmHg but without symptoms, LV systolic dysfunction, LVH, or abnormal exercise test may be considered for repair when the surgical risk is low</td>
<td>IIb</td>
<td>C</td>
</tr>
</tbody>
</table>

ESC Guidelines for the management of grown-up congenital heart disease (new version 2010)
Coarctation of the aorta

- 5 – 8% all congenial heart defects
- Associated with:
  - BAV
  - Subvalvular, supravalvular AS
  - Turner syndrome
  - Shone’s syndrome
  - Congenital rubella syndrome
- Hourglass deformity
- Localized fibrous diaphragm
- Diffuse hypoplasia of descending aorta
CT in diagnosis of CoA
Structural assessment
Doppler examination

Diastolic ‘run-off’ phenomenon is most reliable sign of significant coarctation
<table>
<thead>
<tr>
<th>Indications</th>
<th>Class</th>
<th>Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>All patients with a non-invasive pressure difference &gt; 20 mmHg between upper and lower limbs, regardless of symptoms but with upper limb hypertension (&gt;140/90 mmHg in adults), pathological blood pressure response during exercise, or significant LVH should have intervention</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>Independent of the pressure gradient, hypertensive patients with ≥50% aortic narrowing relative to the aortic diameter at the diaphragm level (on CMR, CT, or invasive angiography) should be considered for intervention</td>
<td>IIa</td>
<td>C</td>
</tr>
<tr>
<td>Independent of the pressure gradient and presence of hypertension, patients with ≥50% aortic narrowing relative to the aortic diameter at the diaphragm level (on CMR, CT, or invasive angiography) may be considered for intervention</td>
<td>IIb</td>
<td>C</td>
</tr>
</tbody>
</table>
Post-operation assessment

- Stent implantation
- Operation
  - End-to end anastomosis
  - Subclavian flap aortoplasty
  - Prosthetic flap aortoplasty

Increased systolic flow rates may developed due to a lack of aortic compliance
Marfane syndrome

- Autosomal, dominant disorder of connective tissue
- Prevelance 2 – 3 per 10 000
- Mutations in the FBN<sup>1</sup> gene encoding fibrilline-1 (glycoprotein in the extracellular matrix)
- Deficiency of fibrillin may lead to weakening of the supportive tissues
- More than 1000 mutations have been identified
- Progressive dilation of the aorta
How to diagnose Marfan syndrome?

- Family history of heart-related death
- Physical features:
  - Skeletal frame (the ratio of arm/leg size to trunk size)
  - Lens dislocation
  - Aortic and valve abnormality
- Genetic analysis (?)
Dilation of the aortic root in Marfan syndrome

Dilation of the aortic root is found in about 60-80% of patients with Marfan syndrome.
Ascending aorta dilation in Marfan syndrome
Marfan syndrome

Progressive dilation of the aorta leads to aortic dissection or rupture

Dissection may occur in patients with mild aortic dilation

Completely safe diameter of the aorta does not exist

Higher risk of re-dissection and reccurent aneurysm
Turner syndrome

- Chromosomal abnormality in which one or part of X chromosome is absent
- Prevalence 1 : 2 500 woman
- Characteristic physical abnormalities (short stature, broad chest, low-set ears, webbed neck)
- Cardiovascular malformations 17 - 45%
  - Bicuspid aortic valve up to 15%
  - Coarctation of the aorta 5 – 12%
  - Aortic dilation and rupture
  - Hypoplastic left heart syndrome
Aortic dilation and dissection in population with CHD

<table>
<thead>
<tr>
<th></th>
<th>Marfan syndrome</th>
<th>Isolated BAV</th>
<th>Coarctation</th>
<th>Turner syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Incidence population</strong></td>
<td>1 : 5 000</td>
<td>&gt; 1 : 100</td>
<td></td>
<td>1 : 2 500 (weman)</td>
</tr>
<tr>
<td><strong>Site of dilation</strong></td>
<td>Aortic root</td>
<td>Asc aorta</td>
<td>Root and asc aorta</td>
<td>Asc aorta</td>
</tr>
<tr>
<td><strong>Age onset, y</strong></td>
<td>20 - 40</td>
<td>50’s</td>
<td></td>
<td>20 - 40</td>
</tr>
<tr>
<td><strong>Progressive dilation</strong></td>
<td>yes</td>
<td>+/-</td>
<td>+/-</td>
<td>?</td>
</tr>
<tr>
<td><strong>Aortic dissection</strong></td>
<td>50%</td>
<td>0.5%</td>
<td>1% (?)</td>
<td>1 – 2%</td>
</tr>
</tbody>
</table>

Curr Opin Cardiol 2008,23,519
“Echocardiography remains the first-line investigation and continues to evolve”