Congenital heart disease

Ewa Szczerba M.D.

18.11.2014
Plan of the seminar

- Embology
- Atrial septal defect (ASD)
- Ventricular septal defect (VSD)
- Atrioventricular septal defect (AVSD)
- Patent ductus arteriosus (PDA)
- Left ventricular outflow tract obstruction (LVOTO)
- Coarctation of aorta (CoA)
- Tetralogy of Fallot (ToF)
- Ebstein’s anomaly
- Marfan syndrome
- Transposition of great arteries (TGA)
Embryology

Source: Congential Malformations of the herat part 1. by R. Rushmer, R Blandau University of Washington
http://www.youtube.com/watch?v=5DIUk9IXUaI
Eisenmenger syndrome

- Caused by increased flow through pulmonary vascular system due to left to right shunt
- Pulmonary hypertension develops
- Reversal of the shunt (right to left)
- Signs and symptoms:
  - Cyanosis
  - High red blood cell count
  - Nail clubbing of fingers
  - Syncope
  - Heart failure symptoms
  - Arrhythmia
Artial Septal Defect (ASD)

• It may remain undiagnosed until adulthood

• Most common types:
  – Ostium primum (15% of ASD)
  – Ostium secundum (80% of ASD)
Clinical presentation

• Majority develop symptoms >40 years old
• Reduced functional capacity
• Shortness of breath on exercise
• Palpitation
• Pulmonary infections
• Signs of right heart failure
Consequences

- Pulmonary vascular disease <5% (measurement of pulmonary vascular resistance PVR)
- Supraventricular tachyarythmias mostly atial flutter or atrial fibrillation
- Systemic embolism:
  - Paradoxical embolism associated with deep vein thrombosis
  - Embolism associated with AF/Afl
Upon clinical examination

• Splitting of the second heart sound in the oscillation area for pulmonary valve
• Systolic pulmonary flow murmur
ECG

Enlarged ‘p’ wave indicating Right atrial hypertrophy

Also note that the aVF is predominantly upwards as compared to Lead I indicating Right Axis Deviation

rSR’ seen and tall R wave indicating RBBB and RVH

LAD with rSR’ in V1 is suggestive of Ostium primum defect
Atrial Septal Defect
Diagnostic Work-up

- **Echocardiography**
  Key diagnostic technique providing diagnosis and quantification (RV volume overload):
  - TEE for precise evaluation of secundum defects prior to device closure (size, residual septal morphology, rim size and quality, exclusion of additional defects and confirmation of normal pulmonary venous connection) and of sinus venosus defects,
  - Other key inform. to be provided includes PAP and TR.

- **CMR/CT**
  If echo is insufficient, particularly for RV volume overload and pulmonary venous connection.

- **Cardiac catheterization**
  Estimation of PVR when echo PAP > 50% of systemic pressure.
Echocardiographic view
“Bubble” test
Indications for Intervention in Atrial Septal Defect

- Patients with significant shunt (signs of RV volume overload) and PVR < 5 WU should undergo ASD closure regardless of symptoms.

- Device closure is the method of choice for secundum ASD closure when applicable.

- All ASDs regardless of size in patients with suspicion of paradoxical embolism (exclusion of other causes) should be considered for intervention.

- Patients with PVR ≥ 5 WU but < 2/3 SVR or PAP < 2/3 systemic pressure (baseline or when challenged with vasodilators, preferably nitric oxide, or after targeted PAH therapy) and evidence of net L-R shunt (Qp:Qs > 1.5) may be considered for intervention.

- ASD closure must be avoided in patients with Eisenmenger physiology.

---

a = class of recommendation. b = level of evidence.

ASD = atrial septal defect; L-R shunt = left-to-right shunt; PAH = pulmonary arterial hypertension;
PAP = pulmonary artery pressure; PVR = pulmonary vascular resistance; Qp:Qs = pulmonary to systemic flow ratio;
SVR = systemic vascular resistance; WU = Wood units.
Atrial Septal Defect
Follow-up

- **FU evaluation** should include assessment of a residual shunt, RV size and function, TR and PAP by echocardiography as well as of arrhythmias by history, ECG and only if indicated (not routinely) Holter.

- Late post-op. **arrhythmias** after surgical repair at age < 40 years are most frequently intraatrial reentrant tachycardia or atrial flutter which can be successfully treated with radiofrequency ablation. With or without repair after 40 years, atrial fibrillation becomes more common (oral anticoagulation!).

- Patients repaired < 25 yrs without relevant sequelae or residuae do not require regular FU; the others should be followed on a regular basis including evaluation in specialized GUCH centers.
**Additional information**

- **Exercise/sports:** No restrictions in asymptomatic patients before or after intervention without pulmonary hypertension, significant arrhythmias, or RV dysfunction.
- **Limitation to low-intensity recreational sports in PAH patients**
- **The risk from pregnancy in patients without pulmonary hypertension is low.**
- **Closure before pregnancy may prevent paradoxical embolism and worsening of clinical status.**
- **Pregnancy is contraindicated in patients with severe PAH or Eisenmenger syndrome**
- **The recurrence rate of CHD is 3–10% (excluding familial ASD and heart–hand syndromes with autosomal dominant inheritance)**
Ventricular septal defect
Ventricular septal defect

- Most common, after bicuspid valve, congenital heart malformation at birth as an isolated finding (30-40%)
- Spontaneous closure is frequent during childhood (in muscular and perimembranous type)
- Mostly diagnosed before adulthood
- Can be a component of complex anomalies such as tetralogy of Fallot.
Types

- Perimembranous (~80%)
- Muscular/trabecular (15-20%)
- Outlet supracristal/subarterial/subpulmonary/infundibular (~5%)
- Inlet/AV canal/AVSD type (typically in Down syndrome)
Clinical presentation

• Depends on the size of the defect, significance of the L-R shunt, LV volume overload, pulmonary hypertension

• If small – no clinical manifestation

• If large – dyspnea on exercise
  – heart failure symptoms
  – Holosystolic murmur over III-IV intercostal space
  – precordial thrill
Problems in adulthood

• Eisenmenger syndrome (in large VSD L-R shunt after development of PAH changes into R-L shunt -> cyanosis)
• Endocarditis 6x higher than in the population
• Heart failure
• Arrythmia
Ventricular Septal Defect Diagnostic Work-up

- **Echocardiography**
  Key diagnostic technique providing diagnosis and quantification (LV volume overload).
  Key findings to provide are location, number and size of defects, severity of LV volume overload and PAP.
  Check for AR due to prolapse of the right or non-coronary cusp (especially in outlet/suprarcristal and high perimembranous VSDs) and for DCRV.

- **CMR**
  If echo is insufficient, particularly for assessment of LV volume overload and shunt quantification.

- **Cardiac catheterization**
  Estimation of PVR when echo PAP > 50% of systemic pressure.
Indications for Intervention in Ventricular Septal Defect

- Patients with symptoms that can be attributed to L-R shunting through the (residual) VSD and who have no severe pulmonary vascular disease (see below) should undergo surgical VSD closure.
- Asymptomatic patients with evidence of LV volume overload attributable to the VSD should undergo surgical VSD closure.
- Patients with a history of IE should be considered for surgical VSD closure.
- Patients with VSD associated prolapse of an aortic valve cusp causing progressive AR should be considered for surgery.
- Patients with VSD and PAH should be considered for surgery when there is still net L-R shunt (Qp:Qs > 1.5) present and PAP or PVR are < 2/3 of systemic values (baseline or when challenged with vasodilators, preferably nitric oxide, or after targeted PAH therapy).
- Surgery must be avoided in Eisenmenger VSD and when exercise induced desaturation is present.
- If the VSD is small, not subarterial, does not lead to LV volume overload or pulmonary hypertension and there is no history of IE, surgery should be avoided.

AR = aortic regurgitation; IE = infective endocarditis; LV = left ventricle; PAH = pulmonary arterial hypertension; L-R shunt = left-to-right shunt; PVR = pulmonary vascular resistance; Qp:Qs = pulmonary to systemic flow ratio; VSD = ventricular septal defect.
Ventricular Septal Defect Follow-up

- **FU evaluation** should include assessment of AR, TR, degree of (residual) shunt, LV dysfunction, elevation of PAP, development of DCRV and development of discrete subaortic stenosis by echocardiography.

- Possible development of complete AV block requires attention (patients who develop bifascicular block or transient trifascicular block after VSD closure are at risk).

- Patients with LV dysfunction, residual shunt, PAH, AR, RVOT or LVOT obstruction should be seen every year, small VSDs in 3-5 year intervals; after device closure: regular follow-up until 2 years and then depending on the result every 2-4 years is recommended. After surgical closure without residual abnormality 5-year intervals.
Additional information

• Exercise/sports: No restrictions are required in patients after VSD closure, or with small VSD without pulmonary hypertension, significant arrhythmias, or LV dysfunction. Patients with PAH must limit themselves to low-intensity recreational activity/sports.

• Pregnancy is contraindicated in Eisenmenger syndrome. The risk is low in asymptomatic patients with normal LV and no PAH.

• The recurrence rate of CHD has been reported at 6–10%.
Atrioventricular septal defect (AVSD)

- Common AV annulus guarded by five leaflets
- ~3% of all CHD
- Associated with Down Syndrome, ToF and other complex CHD
Clinical presentation

- Depends on the presences and size of ASD and VSD and competences of the left-sided AV valve
- Exercise intolerance, dyspnoea, arrythmia, cyanosis
- If unoperated complete AVSD-> Eisenmenger syndrome
Treatment

- Cases of complete AVSD should be operated in 2-4 month of life,
- If symptoms of heart failure unmanageable with medication operation even in 1st month
Atrioventricular Septal Defect (AVSD) Diagnostic Work-up

- **Echocardiography**
  Key diagnostic technique providing assessment of each anatomic component of the AVSD, of the AV-valves and their connections (straddling; overriding) and the severity and exact substrate of AV valve regurgitation, the magnitude and direction of intracardiac shunting, LV and RV function, PAP and the assessment of presence/absence of sub-aortic stenosis.

- **CMR**
  Indicated when additional quantification of ventricular volumes and function or intracardiac shunting is required for decision making.

- **Cardiac catheterization**
  Estimation of PVR when echo PAP > 50% of systemic pressure.
Indications for Intervention in Atrioventricular Septal Defect

Complete AVSD
- Cardiac surgery must be avoided in patients with Eisenmenger physiology. In case of doubt, PVR testing is recommended.
- For indication of intervention see also VSD.

Partial AVSD
- Surgical closure should be performed in case of significant volume overload of the RV. For further details see ASD.

AV valve regurgitation
- Symptomatic patients with moderate to severe AV valve regurgitation should undergo valve surgery, preferably AV valve repair.
- Asymptomatic patients with moderate or severe left-sided valve regurgitation and LVESD > 45 mm and/or impaired LV function (LVEF < 60%) should undergo valve surgery when other causes of LV dysfunction are excluded.
- Surgical repair should be considered in asymptomatic patients with moderate or severe left-sided AV valve regurgitation who have signs of volume overload of the LV and a substrate of regurgitation that is very likely to be amenable for surgical repair.

SubAS
- See LVOTO.

ASD = atrial septal defect; AV = atrioventricular; AVSD = atrioventricular septal defect; LV = left ventricle; LVEF = left ventricular ejection fraction; LVESD = left ventricular end-systolic diameter; PVR = pulmonary vascular resistance; RV = right ventricle; SubAS = subaortic stenosis; VSD = ventricular septal defect.
AVSD
Follow-up

- **FU evaluation** should pay particular attention to residual shunt, AV valve malfunction, LV and RV enlargement and dysfunction, PAP elevation, subaortic stenosis and arrhythmias.

- Life-long regular follow-up of all patients, operated and non-operated, with an AVSD is recommended including evaluation in specialized GUCH centers. The frequency of outpatient visits depends on the presence and severity of residual abnormalities. A surgically repaired AVSD without significant residual abnormalities should be seen at least every 2 to 3 years. In case of residual abnormalities, the intervals should be shorter.
Patent ductus arteriosus (PDA)

- Persistent communication between the proximal left pulmonary artery and the descending aorta just distal to the left subclavian artery
- Isolated or as a part of complex CHD
Clinical presentation

• Small duct with no LV volume overload (normal LV) and normal PAP (generally asymptomatic)

• Moderate PDA with predominant LV volume overload: large LV with normal or reduced function (may present with left heart failure)

• Moderate PDA with predominant PAH: pressure-overloaded RV (may present with right heart failure)

• Large PDA: Eisenmenger physiology with differential hypoxaemia and differential cyanosis (lower extremities cyanotic, sometimes left arm, too);
Patent Ductus Arteriosus (PDA) Diagnostic Work-up

- **Echocardiography**
  Key diagnostic technique providing the diagnosis (may be difficult in pts. with Eisenmenger physiology), the degree of LV volume overload, PAP, PA size and right heart changes.

- **CMR**
  Indicated when additional quantification of LV volumes or evaluation of PA anatomy are required.

- **Cardiac catheterization**
  Estimation of PVR when echo PAP > 50% of systemic pressure.
Indications for Intervention in Patent Ductus Arteriosus

- PDA should be closed in patients with signs of LV volume overload.
- PDA should be closed in patients with PAH but PAP < 2/3 of systemic pressure or PVR < 2/3 of SVR.
- Device closure is the method of choice where technically suitable.
- PDA closure should be considered in patients with PAH and PAP > 2/3 of systemic pressure or PVR > 2/3 of SVR but still net L-R shunt (Qp:Qs > 1.5) or when testing (preferably with nitric oxide) or treatment demonstrates pulmonary vascular reactivity.
- Device closure should be considered in small PDAs with continuous murmur (normal LV and PAP).
- PDA closure should be avoided in silent duct (very small, no murmur).
- PDA closure must be avoided in PDA Eisenmerger or patients with severe PAH and exercise-induced lower limb desaturation.

*a = class of recommendation. b = level of evidence.
L-R shunt = left-to-right shunt; LV = left ventricle; PAH = pulmonary arterial hypertension; PAP = pulmonary artery pressure; PDA = patent ductus arteriosus; PVR = pulmonary vascular resistance; Qp:Qs = pulmonary to systemic flow ratio; SVR = systemic vascular resistance.*
Patent Ductus Arteriosus Follow-up

- **FU evaluation.** Echocardiographic evaluation should include LV size and function, PAP, residual shunt and associated lesions.

- Patients with no residual shunt, normal LV and normal PAP do not require regular follow-up after 6 months.

- Patients with LV dysfunction and patients with residual PAH should be followed with 1-3 years intervals depending on severity, including evaluation in specialized GUCH centers.
Additional information

• Exercise/sports: No restrictions in asymptomatic patients before or after intervention without pulmonary hypertension;

• limitation to low-intensity sports in PAH patients.

• No significantly increased risk for pregnancy in patients without pulmonary hypertension. Pregnancy is contraindicated in patients with severe PAH and Eisenmenger syndrome.
Left ventricular outflow tract obstruction (LVOTO)

- Most common site of obstruction is valvular
- Other possible levels: subvalvular or supravalvular
- Most common cause for congenital valvular aortic stenosis is bicuspid aortic valve
- Patients frequently remain asymptomatic for many years
- Prognosis is good and sudden death is rare in asymptomatic patients with good exercise tolerance, even when stenosis is severe.
- Once symptoms - angina pectoris, dyspnoea, or
- Syncope occur the prognosis deteriorates rapidly.
Valvular AS
Diagnostic Work-up

- **Echocardiography**
  Gold standard for diagnosis, degree of calcification, LV function, LVH and associated lesions. With Doppler echocardiography severity of AS is determined from transvalvular peak velocity ($V_{\text{max}}$), mean gradient and continuity equation calculated effective orifice area.
  
  *TEE* may occasionally be helpful to planimeter AVA.

  *Low dose dobutamine echo* is helpful in low flow AS.

- **Exercise testing**
  In asympt. pts. to confirm asympt. status, evaluate exercise tolerance, blood pressure response and arrhythmias for timing of surgery.

- **CMR/CT**
  Mainly required to quantify aortic dilation.

- **Catheterization**
  Only if echo results are uncertain.
## Diagnostic Criteria for Degree of Aortic Stenosis Severity

<table>
<thead>
<tr>
<th></th>
<th>Mild AS</th>
<th>Moderate AS</th>
<th>Severe AS</th>
</tr>
</thead>
<tbody>
<tr>
<td>( V_{\text{max}} ) (m/sec)*</td>
<td>2.0 - 2.9</td>
<td>3.0 - 3.9</td>
<td>( \geq 4.0 )</td>
</tr>
<tr>
<td>Mean gradient (mmHg)*</td>
<td>&lt; 30</td>
<td>30 - 49</td>
<td>( \geq 50 )</td>
</tr>
<tr>
<td>AVA (cm(^2))</td>
<td>&gt; 1.5</td>
<td>1.0 - 1.5</td>
<td>&lt; 1.0</td>
</tr>
<tr>
<td>AVAi (cm(^2)/m(^2) BSA)</td>
<td>( \geq 1.0 )</td>
<td>0.6 - 0.9</td>
<td>&lt; 0.6</td>
</tr>
</tbody>
</table>

AS = aortic stenosis; AVA = aortic valve area; AVAi = indexed AVA; BSA = body surface area.

\( V_{\text{max}} \) = peak transvalvular velocity.

*at normal transvalvular flow.
Indications for Intervention in Valvular Aortic Stenosis (1)

- Patients with severe AS and any valve related symptoms (AP, dyspnoea, syncope) should undergo valve replacement.
- Asymptomatic patients with severe AS should undergo surgery when they develop symptoms during exercise testing.
- Regardless of symptoms, surgery should be performed when systolic LV dysfunction is present in severe AS (LVEF < 50%), unless it is due to other causes.
- Regardless of symptoms, surgery should be performed when patients with severe AS undergo surgery of the ascending aorta or of another valve or coronary artery bypass grafting.
- Regardless of symptoms, aortic surgery should be considered if the ascending aorta is greater than 50 mm (27.5 mm/m² BSA) and no other indications for cardiac surgery are present.
- Asymptomatic patients with severe AS should be considered for surgery when they present with a fall in blood pressure below baseline during exercise testing.

Class\(^a\)  Level\(^b\)

- **I**  
- **I**  
- **I**  
- **IIa**  
- **IIa**

\(^a\) class of recommendation. \(^b\) level of evidence.

AP = angor pectoris; AS = aortic stenosis; BSA = body surface area; LV = left ventricle; LVEF = ventricular ejection fraction.
Indications for Intervention in Valvular Aortic Stenosis (2)

- Asymptomatic patients with severe AS and moderate-to-severe calcification and a rate of peak velocity progression of ≥ 0.3 m/sec/year should be considered for surgery.
- Patients with moderate AS undergoing coronary artery bypass surgery or surgery of the ascending aorta or another valve should be considered for additional valve replacement.
- Severe AS with low gradient (< 40 mmHg) and LV dysfunction with contractile reserve should be considered for surgery.
- Severe AS with low gradient (< 40 mmHg) and LV dysfunction without contractile reserve may be considered for surgery.
- Asymptomatic patients with severe AS and excessive LV hypertrophy (≥ 15 mm), unless this is due to hypertension, may be considered for surgery.

Class\(^a\)  |  Level\(^b\)
---|---
IIa | C
IIa | C
IIb | C
IIb | C

\(^a\) = class of recommendation. \(^b\) = level of evidence.

AP = angor pectoris; AS = aortic stenosis; BSA = body surface area; LV = left ventricle; LVEF = ventricular ejection fraction.
Valvular Aortic Stenosis
Follow-up

- Lifelong and regular follow-up is required, the intervals depend upon the degree of severity of stenosis. It is also necessary after valve intervention at yearly intervals.

- Echocardiographic evaluation of the aortic valve, LV, mitral valve, PAP and imaging of aortic root to determine progression of valve stenosis and aortic dilation are mandatory.
Coarctation of aorta (CoA)

- It occurs as a discrete stenosis or as a long, hypoplastic aortic segment.
- Typically CoA is located in the area where the ductus arteriosus inserts, and only in rare cases occurs ectopically (ascending, descending, or abdominal aorta).
- CoA accounts for 5–8% of all congenital heart defects.
Coarctation of the Aorta

AO = Aorta
PA = Pulmonary Artery
LA = Left Atrium
RA = Right Atrium
LV = Left Ventricle
RV = Right Ventricle

Oxygen-rich Blood
Oxygen-poor Blood
Clinical presentation

• Signs and symptoms depend on the severity of CoA
• Key symptoms may include headache, nosebleeds, dizziness, tinnitus, shortness of breath, abdominal angina, claudication, leg cramps, exertional leg fatigue, and cold feet.
• The natural course may be complicated by left heart failure, intracranial haemorrhage (from berry aneurysm), IE, aortic rupture/dissection, premature coronary and cerebral artery disease, and associated heart defect
• Screening programme: measurement of blood oxygen saturation on arms and legs
Coarctation (CoA) Diagnostic Work-up

- **Echocardiography**
  Provides information regarding site, structure, and extent of CoA, LV function and LVH, associated cardiac abnormalities, and aortic and supraaortic vessel diameters. Doppler gradients are not useful for quantification. A diastolic “run-off”-phenomenon, is presumably the most reliable sign of significant coarctation or recoarctation.

- **CMR/CT**
  Preferred non-invasive techniques to evaluate the entire aorta, depicting site, extent and degree of the aortic narrowing, the aortic arch, the pre- and post-stenotic aorta, aneurysm and collaterals.

- **Catheterization**
  With haemodynamic assessment is still the “gold standard” for CoA evaluation in many centers.
Indications for Intervention in Coarctation of the Aorta

- All patients with a non-invasive pressure difference > 20 mmHg between upper and lower limbs, regardless of symptoms but with upper limb hypertension (> 140/90 mmHg in adults), pathologic blood pressure response during exercise, or significant LVH should have intervention.

- 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.

- 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.

*a = class of recommendation. b = level of evidence.
CMR = cardiac magnetic resonance; CoA = coarctation of the aorta; CT = computed tomography; LVH = left ventricular hypertrophy.
Coarctation Follow-up

- All pts. require regular follow-up at least every second year including evaluation in specialized GUCH centers. Imaging of the aorta (preferably with CMR) is required. Imaging intervals depend on baseline pathology.

- Residua, sequelae and complications include:
  - arterial hypertension at rest or during exercise,
  - recurring or residual CoA may induce or aggravate systemic arterial hypertension,
  - aneurysms of the ascending aorta or at the intervention site (risk of rupture and death),
  - attention is required for BAV, mitral valve disease, premature CAD, berry aneurysms of the circle of Willis (currently, no routine screening is recommend).
Tetralogy of Fallot (ToF)

• ~10% of all CHD
• four features
  – a non-restrictive VSD
  – overriding aorta
  – Right ventricular outflow tract obstruction
    infundibular, valvular, or (usually) a combination
    of both, with or without supravalvular or branch
    PA stenosis
  – Consequent hiperthrophy of the right ventricle
Clinical presentation

• Childhood:
  – systolic heart murmur,
  – progressive cyanosis (in exercise or at rest)
  – anoxemic attacks due to constriction of arteriosus cone
  – retardation in development
• Unoperated – poor prognosis
• Children undergo first surgical repairs between 6-18 months of life:
  – Total correction
  – Two-step treatment: Blalock-Taussig operation (junction between subclavian artery and pulmonary artery)+total correction afterwards
• 85% of children after surgical treatment have proper exercise tolerance and do not require medication
Ebstein’s anomaly

• abnormally formed and apically displaced leaflets of the tricuspid valve
• The apical displacement of the tricuspid valve means that the right heart consists of an RA, an atrialized portion of the RV, and the remaining functional RV.
• The tricuspid valve is often regurgitant
Normal heart

Right atrium

Tricuspid valve

Ebstein’s anomaly

Atrial septal defect

Right atrium

Displaced tricuspid valve allows blood back into right atrium

Blue and red blood being pumped to all parts of the body instead of just red (Aorta)

Red blood coming back to heart from lungs (PV)

Some blue blood leaks back from right ventricle into large right atrium

Blue blood coming back to heart from lower body (IVC)

Tricuspid valve is low down on the right side of the heart making the right ventricle small

Blue blood pumped across hole; mixes with red blood
Associated anomalies

- shunt at the atrial level [secundum ASD or patent foramen ovale (PFO)]
- accessory pathways [Wolff–Parkinson– White (WPW) syndrome]
- VSD,
- PS, pulmonary atresia,
- ToF,
- CoA,
- mitral valve abnormalities
Clinical presentation

• Mild forms -> asymptomatic
• Severe forms
  – high-grade tricuspid regurgitation
  – Right ventricle dysfunction and failure
  – Cerebral abscesses
  – Paradoxical embolism
  – Pulmonary embolism
  – Tachyarrhythmias
  – Sudden cardiac death
  – Infective endocarditis

• Symptoms:
  – Palpitations, dyspnoea, fatigue, poor exercise tolerance, chest pain, peripheral and/or central cyanosis
## Table 14 Indications for intervention in Ebstein’s anomaly

<table>
<thead>
<tr>
<th>Indications</th>
<th>Class</th>
<th>Level</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Indications for surgery</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Surgical repair should be performed in patients with more than moderate TR and symptoms (NYHA class &gt;II or arrhythmias) or deteriorating exercise capacity measured by CPET</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>• If there is also an indication for tricuspid valve surgery, then ASD/PFO closure should be performed surgically at the time of valve repair</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>• Surgical repair should be considered regardless of symptoms in patients with progressive right heart dilation or reduction of RV systolic function and/or progressive cardiomegaly on chest X-ray</td>
<td>IIa</td>
<td>C</td>
</tr>
<tr>
<td><strong>Indications for catheter intervention</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Patients with relevant arrhythmias should undergo electrophysiologic testing, followed by ablation therapy, if feasible, or surgical treatment of the arrhythmias in the case of planned heart surgery</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>• In the case of documented systemic embolism probably caused by paradoxical embolism, isolated device closure of ASD/PFO should be considered</td>
<td>IIa</td>
<td>C</td>
</tr>
<tr>
<td>• If cyanosis (oxygen saturation at rest &lt;90%) is the leading problem, isolated device closure of ASD/PFO may be considered but requires careful evaluation before intervention (see text)</td>
<td>IIb</td>
<td>C</td>
</tr>
</tbody>
</table>
Marfan syndrome

- Autosomal dominant disorder of connective tissue
- Involvement: cardiovascular, skin and skeletal, ocular, pulmonary, and dura mater
- Prevalence ~2–3 per 10 000 births
- Mutations in the FBN1 gene on chromosome 15q21 encoding fibrillin-1, a glycoprotein in the extracellular matrix
Clinical presentation

- Long arms, legs and fingers
- Tall and thin body type
- Curved spine
- Abnormalities of the chest
- Flexible joints
- Flat feet
- Crowded teeth
- Stretch marks on the skin that are not related to weight gain or loss

- Prognosis is mainly determined by progressive dilation of the aorta, leading to aortic dissection or rupture, which are the major causes of death
- Men age of untreated patients: 40 years old.
Medical therapy

• Both medical and surgical therapies have improved life expectancy substantially up to 60 – 70 years.
• Blockers might reduce the rate of aortic dilation and might improve survival, at least in adults.
• Rigorous antihypertensive medical treatment, aiming at a systolic blood pressure, 120 mmHg, and 110 mmHg in patients with aortic dissection, is important.
### Indications for surgery in MS

#### Table 12: Indications for aortic surgery in Marfan syndrome

<table>
<thead>
<tr>
<th>Indications</th>
<th>Class</th>
<th>Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patients should undergo surgery when aortic root maximal diameter is:</td>
<td>I</td>
<td>C^c</td>
</tr>
<tr>
<td>* &gt;50 mm</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• 46–50 mm with</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>- family history of dissection or</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>- progressive dilation &gt;2 mm/year as confirmed by repeated measurement or</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>- severe AR or MR or</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>- desire of pregnancy</td>
<td>I</td>
<td>C</td>
</tr>
<tr>
<td>• Patients should be considered for surgery when other parts of the aorta &gt;50 mm or dilation is progressive</td>
<td>IIA</td>
<td>C</td>
</tr>
</tbody>
</table>

^a Class of recommendation.  
^b Level of evidence.  
^c ESC guidelines for valvular heart disease are slightly more strict, recommending only one diameter (45 mm) regardless of other findings.  
AR = aortic regurgitation; MR = mitral regurgitation.
Additional information

• Patients should be advised to avoid exertion at maximal capacity, competitive, contact, and isometric sports.
• There is a 50% chance that a child born to a mother with Marfan syndrome would be affected with the condition (genetic counselling).
• Women with an aortic diameter >45 mm are strongly discouraged from becoming pregnant without prior repair because of the high risk of dissection.
• An aortic diameter <40 mm rarely presents a problem, although a completely safe diameter does not exist.
• With an aorta between 40 and 45 mm, previous aortic growth and family history are important for advising pregnancy with or without aortic repair.
• Even after repair of the ascending aorta, Marfan patients remain at risk for dissection of the residual aorta.
Transposition of great arteries (TGA)

• characterized by ventriculo-arterial discordance: the LV gives rise to the PA, and the RV to the aorta

• A complex TGA associates with intracardiac anomalies:
  – VSD (in up to 45% of cases),
  – LVOTO (~25%),
  – CoA (~5%)
After birth

• Requires either isoprostin admision to keep the ducus arteriosus open
• ENSURE the connection between the two circulations!
• Treatment:
  – Mustard or Senning atrial switch procedure
  – Arterial switch procedure
Thank you for your attention