Ποιες συγγενείς παθήσεις των κολποκοιλιακών βαλβίδων εκδηλώνονται αρχικά στην ενήλικη ζωή και ποια η ενδεδειγμένη αντιμετώπισή τους;

Σεμινάρια Ομάδων Εργασίας
Θεσσαλονίκη
21-23 Φεβρουαρίου 2019

Μπούτσικου Μαρία MD, MSc, PhD
Υπ. Τμήματος Μαγνητικού καρδιακού Συντονισμού, Νοσοκομείο Mediterraneo, Αθήνα
ACHD/MRI Unit, Royal Brompton Hospital, London, UK
No conflicts of interest
Mitral valve prolapse (MVP)

- It is the most common cardiac valvular anomaly in developed countries.
- MVP occurs when the leaflets extend above the plane of the mitral annulus during ventricular systole at least 2 mm beyond the long-axis annular plane, with or without a thickening of leaflets.
- The most serious complication is severe mitral valve regurgitation.
- Mitral valve repair is recommended in patients with symptomatic severe mitral regurgitation or in asymptomatic patients with ventricular enlargement or dysfunction.
Mitral valve in Marfan Syndrome

- MVPs associated with Marfan syndrome ranges from 40 to 91%. Caused by myxomatous degeneration and proliferation of mitral valve collagen best recognized by electron microscopy.
- Marfan syndrome is associated with mutations in fibrillin-1 and in TGF-receptor 2.
- The degree of severity ranges from mild isolated billowing of the valve leaflets to pronounced prolapse with severe valvular insufficiency.
- Commonly involves both leaflets and is symmetrical in Marfan syndrome, whereas it more frequently affects one leaflet (posterior) in myxomatous degeneration.
- Mitral valve regurgitation, requiring surgery, preferably repair, when hemodynamically important.
Indications for Aortic Surgery in Marfan Syndrome

- Patients should undergo surgery when aortic root maximal diameter is:
  - > 50 mm
  - 46-50 mm with:
    - family history of dissection or,
    - progressive dilation > 2 mm/year as confirmed by repeated measurement or,
    - severe AR or MR or,
    - desire of pregnancy.

- Patients should be considered for surgery when other parts of the aorta > 50 mm or dilation is progressive.

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

* ESC guidelines for valvular heart disease are slightly more strict, recommending only one diameter (45 mm) regardless of other findings. In small individuals, the use of an indexed diameter adjusted for BSA of 2.75 cm/m² should probably be used.

AR = aortic regurgitation; MR = mitral regurgitation.
Isolated cleft of the anterior MV leaflet

- Rare but well-known finding
- Division of one of the leaflets (usually the anterior leaflet) of the mitral valve.
- Should not be mistaken with the so-called ‘cleft’ in AVSD
- Classified into two distinct groups: cleft with normally related great arteries and cleft with abnormal conus associated with TGA and DORV
- In AVSD, the positions of both papillary muscles were rotated counterclockwise, whereas in isolated cleft, the position of the papillary muscles was similar to that in normal patients.
- Severe MR in 50% of cases
- MV repair is preferred to MVR and usually consists of a direct suture of the cleft.
- When surgical treatment is performed in adults, the edges of the leaflets tend to be thicker and more retracted, which makes the repair more complicated.
AVSD anatomy

- An AVSD is characterized by the presence of a common AV annulus, guarded by five leaflets.
  - Partial form, the anterior and posterior bridging leaflets are fused centrally, creating separate left- and right-sided orifices.
  - Complete AVSD the central fusion is not present and there is only one orifice.

- AVSD commonly occurs as part of more complex CHD, extracardiac defects and genetic syndromes.
  - ~50% Down syndrome.
  - Tetralogy of Fallot.
Recognizing the level of shunt

Levels of shunting:

A. Interatrial
B. Interatrial and interventricular
C. Interventricular
Assess anatomy

AVSD with two separate AV valves orifice. No ventricular component. Cleft of the mitral valve
Other morphologic features

• The common AV valve leaflets may be dysplastic / functionally regurgitant or stenotic.
• Left AV valve regurgitation or stenosis may affect outcome
• The common AV valve and junction may be dominantly committed to one or other ventricle, resulting in marked ventricular disproportion so that one ventricle is too small to support the circulation.
Clinical presentation and natural history

• Outcome mainly depend on the presence and size of the ASD and VSD and competence of the left-sided AV valve.

• Symptoms are not specific are caused by intracardiac shunting (L–R, R–L, or bidirectional), pulmonary hypertension, AV valve regurgitation, ventricular dysfunction, or LVOTO, sub AS.

• Unoperated complete AVSD \( \rightarrow \) Eisenmenger syndrome unless the VSD is only small.
Clinical presentation in adulthood

Adults with an unoperated AVSD would normally fit into one of the following three diagnostic groups:

• Partial AVSD or primum ASD (with or without a small ventricular component)- benefit from surgery

• Complete AVSD with RVOT (like ToF)—should be considered for surgery

• Complete AVSD with irreversible pulmonary vascular disease (Eisenmenger physiology)—diagnosis needs to be confirmed; conventional surgery is not an option but patient should be considered for anti-pulmonary hypertensive therapy
Therapeutic options

• Partial AVSD (primum ASD) with right-sided heart dilation for elective repair irrespective of symptoms transcatheter closure is not an option, hence operation is required.
• The left AV valve needs to be assessed and is usually operated on at the time to ensure its competence and avoid stenosis.
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.

Class: IIa, II, III; Level: C, B, A, B, C

European Heart Journal 2010; doi:10.1093/eurheartj/ehq249
Congenital Mitral Stenosis

- Rare anomaly occurring in 0.6% of congenital heart disease
- A variety of anatomic features may be present.
  - Parachute mitral valve: single papillary muscle is present to which all the mitral chordae attach;
  - Two closely spaced papillary muscles with an arcade of short chordae limiting leaflet mobility;
  - A supravalvular mitral ring in which a fibrous membrane is present on the left atrial side of the mitral valve, frequently adherent to the mitral leaflets.
- The most common association is with other left-sided anomalies, particularly obstructive lesions.
  - With CoA, BAV, and subaortic stenosis.
  - VSD, ASD, ToF

Gatzoulis et al. Diagnosis and Management of Adult Congenital Heart Disease
Archives of Cardiovascular Disease (2011) 104, 465—479
Parachute mitral valve

- All chordae tendineae insert into a single papillary muscle head, forming a parachute like deformity.
- A second hypoplastic papillary muscle may be present but does not receive chordae tendineae.
- In patients with AVSD the antero-lateral papillary muscle is usually dominant.
In 1963 Shone described a constellation of defects including:

- Parachute mitral valve
- Discrete subaortic stenosis
- Aortic coarctation
- The supramitral ring: fibrous membrane originating just above the mitral annulus, beneath the orifice of the left atrial appendage not adhering to the leaflets and associated with a normal subvalvular apparatus.
- Must be distinguished from cor triatriatum sinister, which is a fibromuscular membrane, clearly separated from the mitral valve that divides the left atrium into two parts.

Double orifice mitral valve

- Rare congenital cardiac anomaly (>200 cases have been reported).
- Associated with AVSD, trisomy 21, Ebstein’s anomaly, CoA and BAV
- DOMV consists of two anatomically distinct mitral orifices that are divided by an accessory bridge of fibrous connective tissue. The fibrous ridge is composed of the mitral leaflet tissue and chordae tendineae.
- The DOMV tensor apparatus is always abnormal. Each orifice is separately attached to their individual, single papillary muscle, and this creates a kind of double parachute mitral valve.
- DOMV has recently been reported on in adulthood or even in the elderly population.
- Clinical implications depend upon the severity of the regurgitation and also on the presence of any stenosis or associated malformations.
Anomalies of the MV apparatus

- Although mitral arcade is not an anomaly of the papillary muscles, it may be seen in association with PMV.
- Is believed to be the result of an arrest in the developmental stage of the mitral valve before attenuation and lengthening of the collagenized chordae tendinae.
- Echo: short chordae and restricted motion of the leaflets with limited coaptation, multiple jets through the reduced interchordal spaces
- MR progressively gets worse, with or without MS
- The valve may function relatively normally for many years.
- Conservative surgery will create two separated papillary muscles by resection of the muscular band.
Presentation in adulthood

- Unoperated congenital mitral stenosis in the adult will be limited to mild abnormalities of the mitral valve or support apparatus with or without mild obstruction.
- A few individuals may survive to adulthood with moderately severe mitral stenosis, particularly if they have not had access to medical care.
- The presenting symptoms and complications should be identical to those in the adult with rheumatic mitral valve disease.
- In addition, these individuals may have other associated congenital abnormalities.
- Occasionally, an adult patient with double-inlet ventricle and intact atrial septum will present with left atrial hypertension due to mitral stenosis.
- This situation, which is treated by atrial septectomy, can lead to left atrial thrombus formation, atrial arrhythmias, and pulmonary hypertension.
Therapeutic options

• Management of the adult with congenital mitral stenosis, includes the initial evaluation, addressing medical management issues and the need for surgical or catheter intervention.

• Because hemodynamically significant congenital mitral stenosis is rare in adults, there is little experience with balloon valvuloplasty but the general view is that balloon valvuloplasty is less effective in patients with congenital mitral stenosis.
Hypertrophic cardiomyopathy

- Commonest genetic cause of heart disease (1:500) (Autosomal Dominant- mutations in genes encoding sarcomere proteins)
  - 90% of pts have familial disease
  - 10% de novo mutations
- Most frequent cause of SCD in young people.
- Increased risk heart failure (older patients), arrhythmias and stroke
- Characterized by myocyte disarray, hypertrophy, and interstitial fibrosis
- 1st degree-relatives -1:2 risk gene carrier
HCM- definition and diagnosis

• Apart from LVH the disease phenotype also includes
  • Electrocardiographic abnormalities
  • Morphologic abnormalities of the MV apparatus
  • Different degree of LVOT obstruction
  • Coronary microvascular abnormalities
  • Myocardial fibrosis (patchy/diffuse)
Morphological findings

- Increased length of MV leaflets-LVOTO
- Fragmented-hypertrophied papillary muscles
- Multiple myocardial crypts

Papillary muscles

Mitral valve

Crypts

Harrigan AJC 2008;101:668

Maron Circ 2011;124:40

Germans JACC 2006;48:2518
PAPILLARY FIBROELASTOMAS

• Papillary fibroelastomas are small pedunculated tumors located on the cardiac valves.

• Typical appearance: short attaching pedicle, resembling a “sea anemone.” Approximately 50% have a stalk; and when they do, they are always mobile.

• More frequently located on the aortic and mitral valves.

• Microscopically, each frond is formed of a central fibroelastic core, an overlying myxomatous layer, and an endothelial covering.

• Large papillary fibroelastomas should be excised owing to the relatively high incidence of embolization. Survival is excellent after surgical excision without tumor recurrence.
Ebstein anomaly

• Wide variation of abnormalities that include
  – Failure of tricuspid valve delamination (adherence of the leaflets to the underlying myocardium)
  – Apical and posterior (downward) displacement of the functional tricuspid annulus
  – Dilation of the “atrialized” portion of the right ventricle
  – Fenestrations, redundancy, and tethering of the anterior leaflet;
  – Dilation of the right atrioventricular junction

– The tricuspid leaflets are dysplastic and are tethered by short chordae and papillary muscles or attached to the underlying myocardium directly by muscular bands. Chordae may be few to absent, and leaflet fenestrations are common.

• The malformed tricuspid valve is usually incompetent

• Most patients in New York Heart Association (NYHA) classes I and II can be managed medically.

• May remain undetected until late childhood or adulthood.

• The most common presenting symptom is fatigue in both older children and adults.
Indications for Intervention in Ebstein’s Anomaly

Indications for surgical repair
- In patients with more than moderate TR and symptoms (NYHA class > II or arrhythmias) or deteriorating exercise capacity measured by CPET.
- If there is also an indication for tricuspid valve surgery, then ASD/PFO closure should be performed surgically at the time of valve repair.
- 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.

Indications for catheter intervention
- 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.
- In the case of documented systemic embolism likely caused by paradoxical embolism, isolated device closure of ASD/PFO should be considered.
- If cyanosis (oxygen saturation at rest < 90%) is the leading problem, isolated device closure of ASD/PFO may be considered but requires careful evaluation before intervention.

a = class of recommendation. b = level of evidence.
ASD = atrial septal defect; CPET = cardiopulmonary exercise testing; NYHA = New York Heart Association; PFO = patent foramen ovale; RV = right ventricle; TR = tricuspid regurgitation; VSD = ventricular septal defect.

European Heart Journal 2010; doi:10.1093/eurheartj/ehq249
Congenital TS/TR

- Isolated congenital TS is rare and presents exclusively in infancy and childhood.

- The typical presentation of adults with TR includes
  - dyspnea,
  - exercise intolerance,
  - palpitation,
  - atrial fibrillation,
  - heart failure.
  - cyanosis should alert one to the possibility of concomitant atrial shunt such as a patent foramen ovale or atrial septal defect with a secondary right-to-left shunt.

- Unoperated patients with severe isolated congenital TR should be considered for TV repair or replacement

- if they have one of the following indications:
  - Symptoms of dyspnea, palpitation, or fatigue thought to be due to TR
  - A patent foramen ovale causing resting or exercise-induced oxygen desaturation
Thank you