Adult congenital heart disease

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ESC
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ESC GUIDELINES

2020 ESC Guidelines for the management of adult congenital heart disease

The Task Force for the management of adult congenital heart disease of the European Society of Cardiology (ESC)

- Congenital heart disease (CHD):
  - Anomaly of the cardiovascular structure or function, which is present at birth, even it is diagnosed later in life

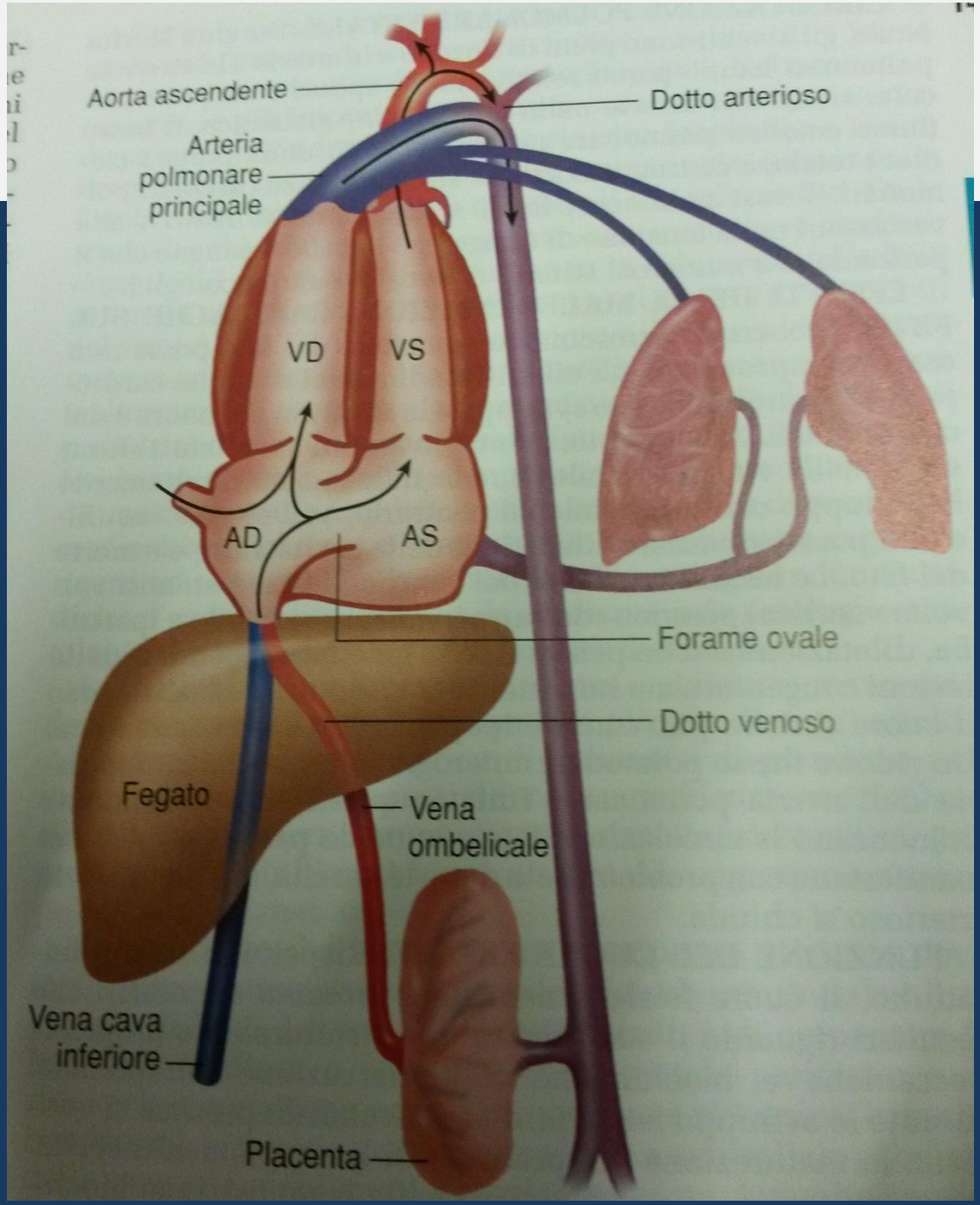
- Pathophysiology:
  - Altered embyogenic development of a normal structure
  - Premature developmental arrest of a structure at an early embryogenic or fetal stage
  - Alterations of the blood flow due to an anatomical defect (e.g. hypoplastic LV due to mitral stenosis)

Fetal circulation

Circulation in-parallel

Oxygenated blood:
placenta → umbilical vein → venous duct (bypassing hepatic microcirculation) → inferior vena cava → patent foramen ovale → systemic circulation

Pulmonary circulation in the fetus equals about 7-10% of the total cardiac output due to high PVR
De-oxygenated blood from RV to systemic circulation through patent ductus arteriosus



- Incidence:
  - 0.8% of alive newborn
  - 50-60% diagnosed within the first 4 weeks of life
- NB thanks to progress in antenatal and pediatric cardiology, the number of patients with congenital abnormalities (completely or partially corrected, as well as non-corrected) will increase over time

etiology

- Environmental factors:
  - Toxics or infectious agents during pregnancy
    - Rubella syndrome
    - Alcohol
    - Drugs
      - thalidomide
      - litium

- Genetics (10-30% of cases) :
  - Single mutation, with or without syndromic manifestations, e.g. :
    - Down's syndrome
    - DiGeorge's syndrome
    - Noonan's syndrome
  - Multifactorial, complex etiology

- Identification of the (main) defect, corrected or uncorrected (and associated abnormalities)

  CLINICAL HISTORY
  ECHO
  CT
  MR

- Identification of the hemodynamic consequences of the defect
  - LV volume overload,
  - RV volume overload,
  - PH
  - L-R, bidirectional, R-L shunt

  ECHO
  MR
  CARDIAC CATH

- Impact on functional capacity

  CPET

Classifications

- Anatomical classification
- Pathophysiological classification
- Classification according to CHD complexity

Anatomical classification

- Septation defects
- Connections abnormalities: veins to atria, atria to ventricles, ventricles to arteries
- (RV or LV) outflow or inflow obstruction
- Any association of the above-mentioned abnormalities
- Viscero-atrial situs abnormalities: cardiac malpositions, often associated to the above-mentioned abnormalities

Pathophysiological classification
Presence or absence of cyanosis (oxygen desaturation)

- Non-cyanotic CHD
  - With L-R shunt
  - Without L-R shunt

- Cyanotic CHD (with R-L shunt)
  - With pulmonary vascular disease (Eisenmenger's syndrome)
  - With pulmonary overflow
    - Complete transposition of the great vessels
    - Total anomalous pulmonary venous connection
  - With normal/low pulmonary flow
    - Ebstein's anomaly
    - Tricuspid atresia
    - Tetralogy of Fallot

Eisenmenger's syndrome

- Pulmonary vascular disease secondary to pre-existing large L-R shunt leading to pulmonary overflow.
- Over time, pulmonary artery pressure and PVR become suprasystemic with bidirectional or R-L shunt
- CHD potentially leading to Eisenmenger's syndrome:
  - Simple defects (ASD, VSD, PDA)
  - Complex defects (AVSD, truncus arteriosus, aorto-pulmonary window, univentricular heart)
- PVR increases during childhood (except in ASDs)

Cyanotic CHD
Systemic clinical manifestations

- Pathophysiology
  - R-L shunt → hypoxemia → renal erythropoietin production → secondary, medullary and extramedullary erythropoiesis → polycytemia

- Clinical consequences
  - Hyperviscosity syndrome: headache, fatigue, vertigo, mental status alterations, visual disturbances, paresthesia, tinnitus, muscle pain
  - Hemostasis abnormalities with predisposition to bleeding
  - Central nervous system
    - Cerebral hemorrage
    - Cerebral emboli (paradoxical embolism through a R-L shunt)
  - Hypertrophic osteopathy
    - Megakaryocytes bypass the pulmonary filter due to R-L shunt and are trapped in peripheral capillaries → release of growth factors → periostitis with pain

Figure 7: Hypertrophic osteoarthropathy in male 26 year old, Eisenmenger syndrome and unrepaired ventricular septal defect, Saturation 70% on room air.

Classification according to CHD complexity

- Mild complexity (→ no specialistic follow-up in most cases)
- Moderate complexity
- Severe complexity

Simple CHD

- Isolated congenital aortic valve disease and bicuspid aortic disease
- Isolated congenital mitral valve disease (except parachute valve, cleft leaflet)
- Mild isolated pulmonary stenosis (infundibular, valvular, supravalvular)
- Isolated small IASD, IVSD, or PDA
- Repaired secundum ASD, sinus venosus defect, VSD or PDA without residuae or sequellae, such as chamber enlargement, ventricular dysfunction, or elevated PAP

ASD=atrial septal defect

IASD=interatrial septal defect

PDA=patent ductus arteriosus

VSD=ventricular septal defect

CHD of moderate complexity

- Anomalous pulmonary venous connection (partial or total)
- Anomalous coronary artery arising from the PA
- Anomalous coronary artery arising from the opposite sinus
- Aortic stenosis – subvalvular or supravalvular
- AVSD, partial or complete, including primum ASD (with no pulm vasc disease)
- ASD secundum, moderate or large or unrepaired (with no pulm vasc disease)
- Coarctation of the aorta
- Double chambered right ventricle
- Ebstein anomaly
- Marfan syndrome and related HTAD, Turner syndrome
- PDA, moderate or large unrepaired (with no pulm vasc disease)
- Peripheral pulmonary stenosis
- Pulmonary stenosis, infundibular, valvular or supravalvular, moderate or severe
- Sinus of Valsalva aneurysm/fistula
- Sinus venosus defect
- Tetralogy of Fallot, repaired
- Transposition of the great arteries after arterial switch operation
- VSD with associated abnormalities (no pulm vasc dis) and/or >= moderate shunt

CHD of severe complexity

- Any CHD (repaired or unrepaired) associated with pulm vasc disease
- Any cyanotic CHD (unoperated or palliated)
- Double-outlet ventricle
- Fontan circulation
- Interrupted aortic arch
- Pulmonary atresia
- Transposition of the great arteries (except arterial switch operation)
- Univentricular heart
- Truncus arteriosus
- Other complex abnormalities of AV and ventriculo-arterial connection

Atrial septal defects

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Department of Management, Information and Production Engineering

According to anatomical localization of the defect, we describe

- Secundum ASD
  - 80% of ASDs
  - Region of the fossa ovalis and its sorroundings
- Primum ASD
  - 15% of ASDs
  - associated with malformation of AV valves, resulting in regurgitation
- Sinus venosus defect
  - Superior sinus venosus defect
    - 5% of ASDs
    - Located near the superior vena cava (SVC) entry
    - Associated with partial or complete connection of right pulmonary vein to SVC / right atrium)
  - Inferior sinus venosus defect
    - < 1% of ASD
    - Located near the inferior vena cava (IVC) entry
- Unroofed coronary sinus
  - < 1% of ASD
  - Separation from the left atrium (LA) can be partially or completely missing

Potentially associated lesions: anomalous pulmonary venous connections, persistent left SVC, pulmonary valve stenosis, mitral valve prolapse

Atrial septal defects

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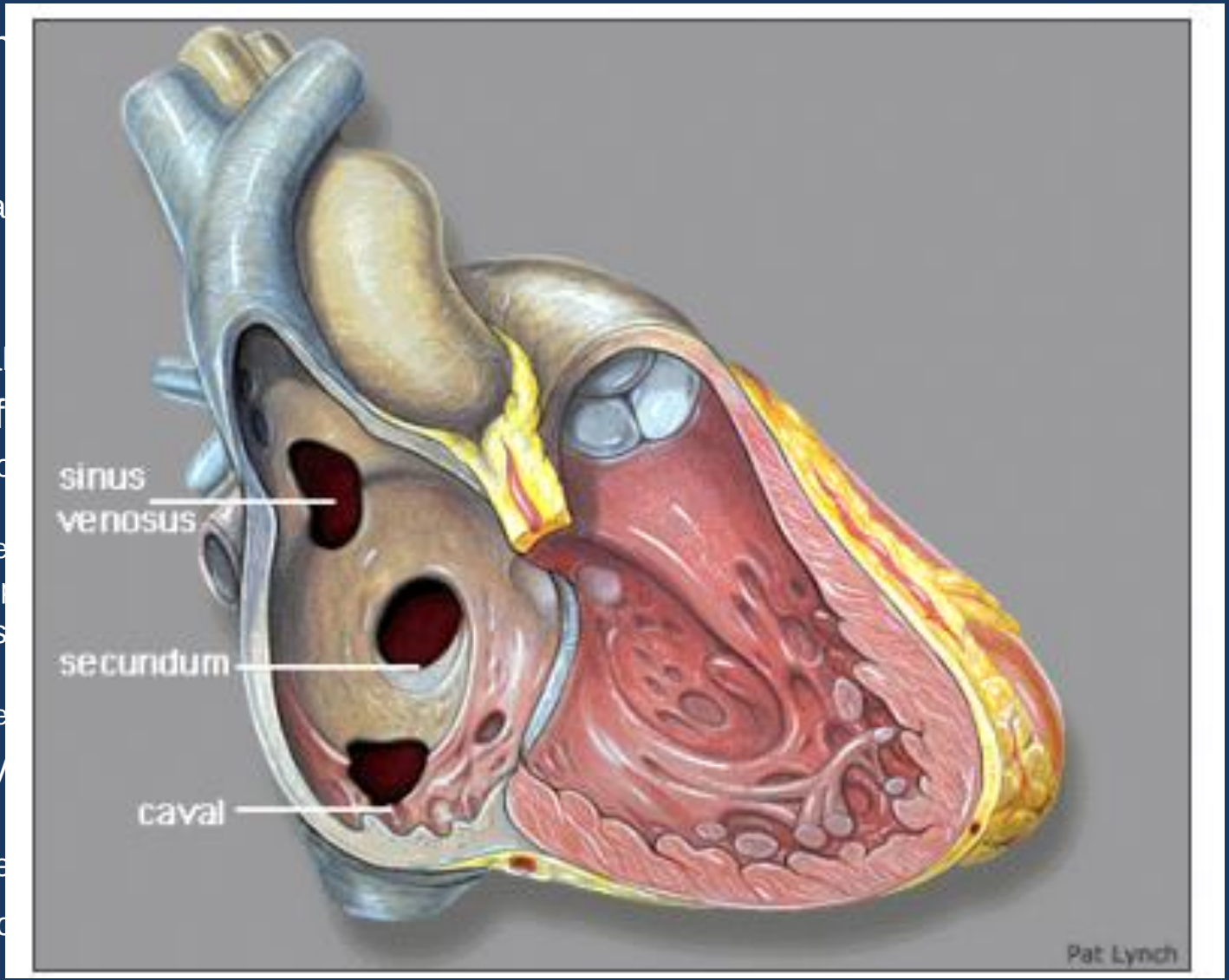
  - Separation from the

Potentially associated lesio
valve stenosis, mitral valve



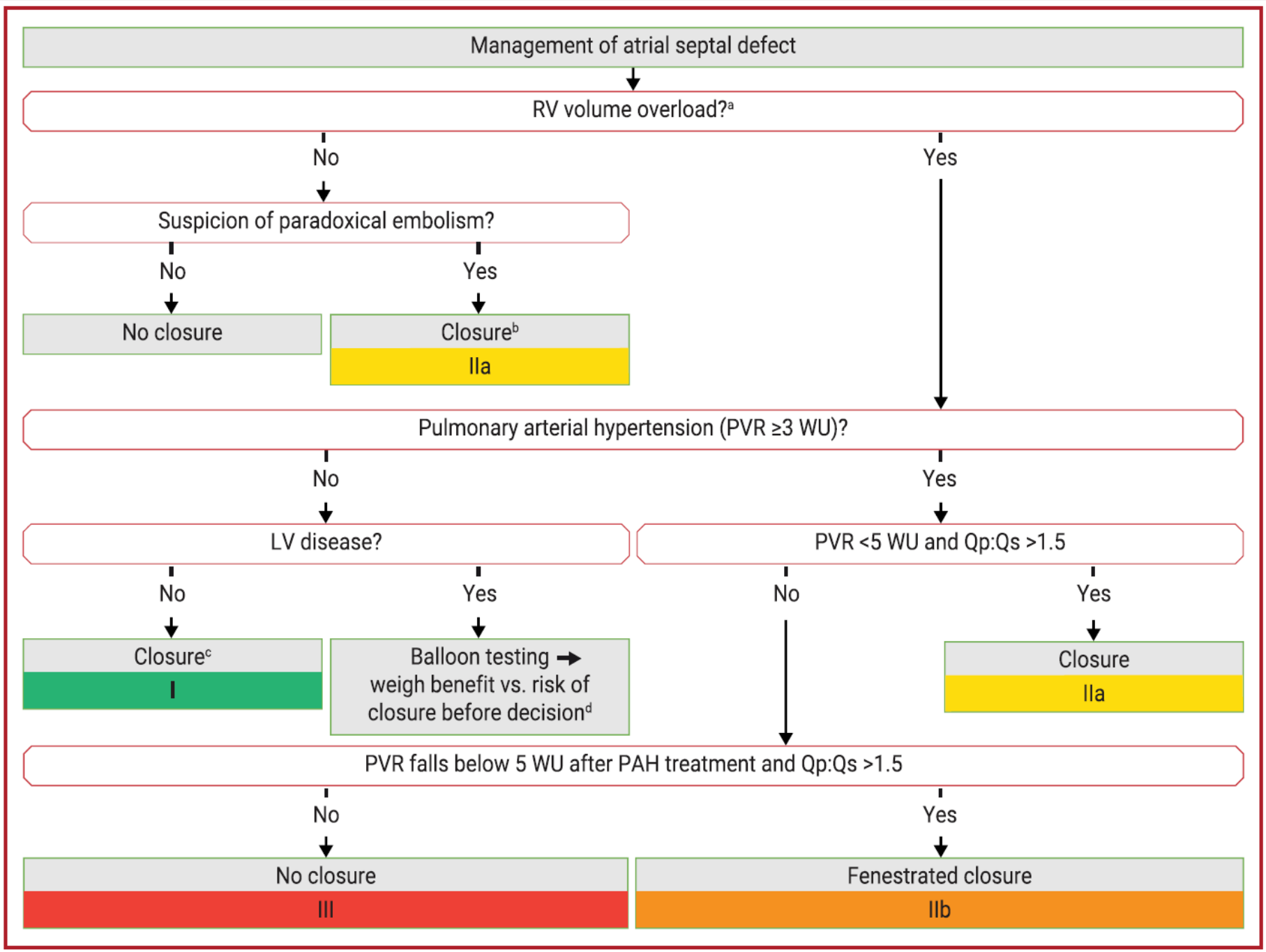
ASDs

- Shunt volume depends on RV/LV compliance, defect size, LA/RA pressure
- A simple ASD results in L-R shunt because of the higher compliance of the RV/RA compared with the LV/LA, leading to higher LA than RA pressure
- Relevant shunt occurs in general with defect size ≥ 10 mm in diameter
- Shunt causes RV volume overload and pulmonary overcirculation
- Severe PVD is rare (<5%) and its development presumably requires additional factors

ASDs

- Generally diagnosed in the newborn or during pediatric age (systolic murmur)

- Generally asymptomatic up to 30-40 years (or even more, depending on ASD dimensions) → reduced functional capacity, exertional shortness of breath, palpitations

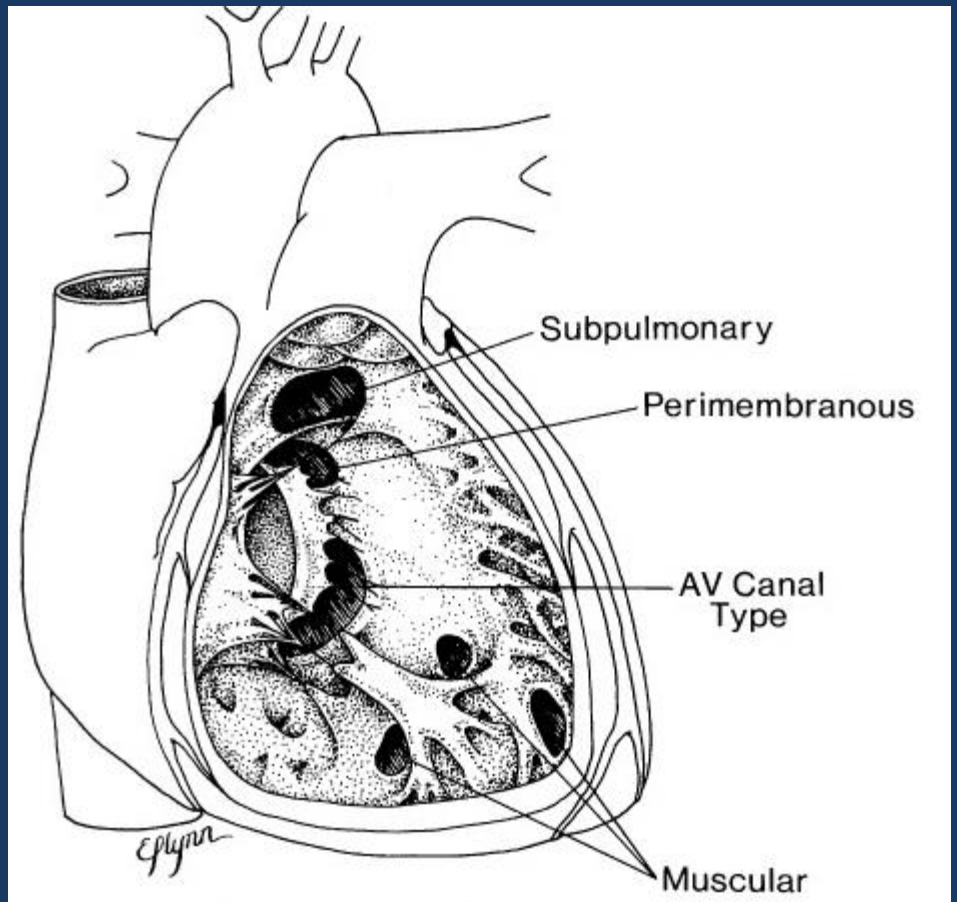

Management of atrial septal defect
RV volume overload?[a]
No
Yes
Suspicion of paradoxical embolism?
No
Yes
No closure
Closure[b]
IIa
Pulmonary arterial hypertension (PVR ≥3 WU)?
No
Yes
LV disease?
PVR <5 WU and Qp:Qs >1.5
No
Yes
No
Yes
Closure[c]
I
Balloon testing → weigh benefit vs. risk of closure before decision[d]
Closure
IIa
PVR falls below 5 WU after PAH treatment and Qp:Qs >1.5
No
Yes
No closure
III
Fenestrated closure
IIb

Ventricular septal defects

IVSDs represent about 35% of CHD

IVSD are classified based on the localization of the defect:

- Perimembranous (80%)
- Muscular/trabecular (15-20%)
- Subpulmonary (5%)
- AV canal type (Down syndrome)



septal defects when viewed from the right ventricle.

VSDs

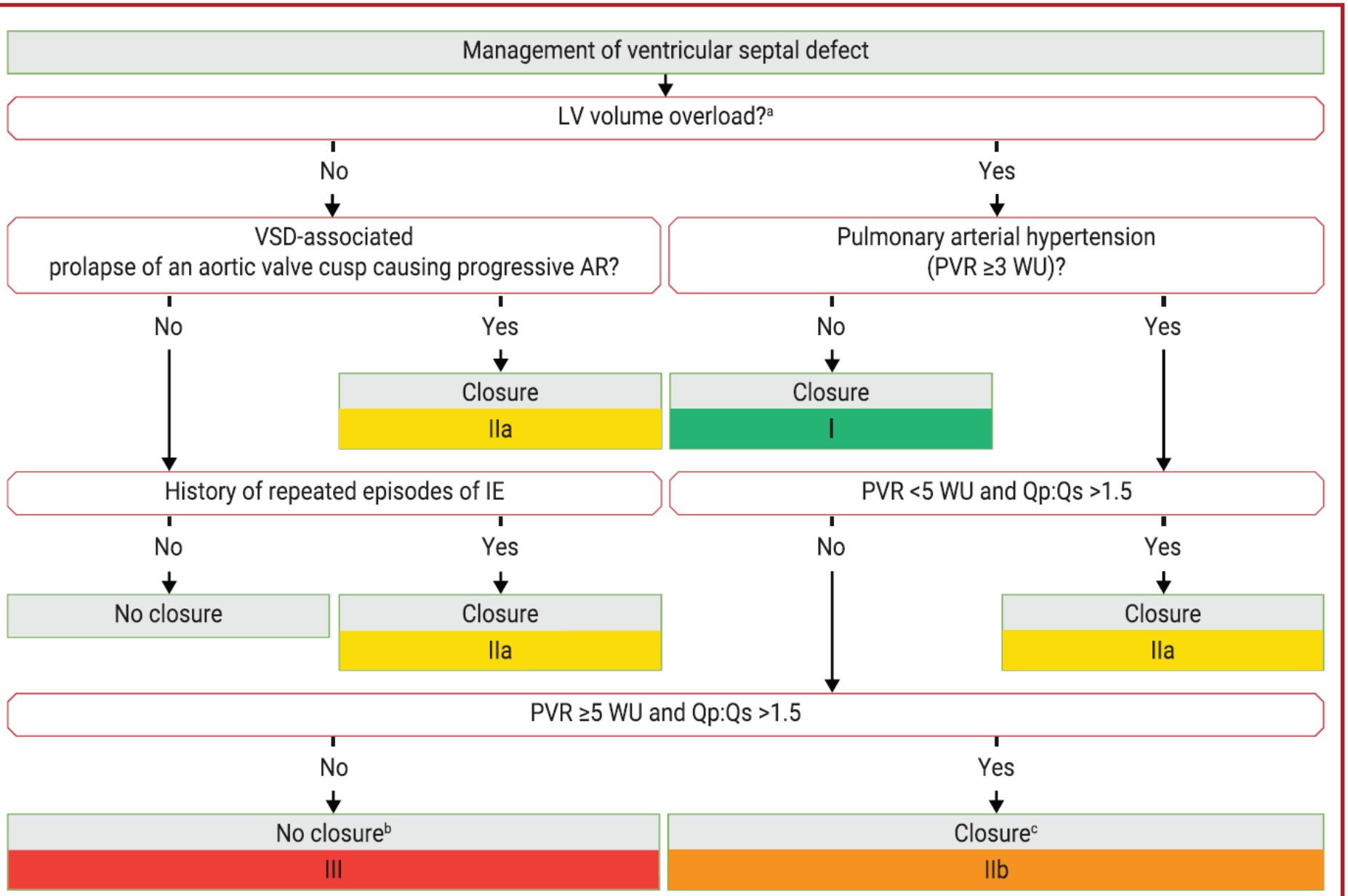
- VSD is mostly diagnosed and – when indicated – treated before adulthood

- Spontaneous closure of small defects is frequent in childhood, especially in perimembranous and muscular defects, thanks to myocardial muscular growth

- The direction and magnitude of the shunt are determined by PVR and systemic vascular resistance, the size of the defect, LV/RV systolic and diastolic function, and the presence of RV or LV outflow tract obstruction

VSDs

- Usual presentation in adulthood include:
  - VSD operated in childhood, without residual VSD and no PH
  - VSD operated in childhood, with residual VSD. The residual shunt size determines the degree of LV volume overload and the development of PH
  - Small VSD with insignificant L-R shunt, without LV volume overload or PH (restrictive VSD), which was not considered for surgery in childhood
  - VSD with L-R shunt, PH, and various degrees of LV volume overload (rare)
  - VSD with R-L shunt (Eisenmenger syndrome): large, non-restrictive VSD, with originally large L-R shunt and development of PVD eventually resulting in shunt reversal


Management of ventricular septal defect
LV volume overload?[a]
No
Yes
VSD-associated prolapse of an aortic valve cusp causing progressive AR?
Pulmonary arterial hypertension (PVR ≥3 WU)?
No
Yes
No
Yes
Closure
IIa
Closure
I
History of repeated episodes of IE
PVR <5 WU and Qp:Qs >1.5
No
Yes
No
Yes
No closure
Closure
IIa
Closure
IIa
PVR ≥5 WU and Qp:Qs >1.5
No
Yes
No closure[b]
III
Closure[c]
IIb

Arrhythmias in CHD are frequent

	Supraventricular arrhythmias			Ventricular arrythmias and SCD		Bradycardia			
Type of CHD	AVRT	IART/ EAT	AF	Sustained VT	SCD	SND		AV block	
						Congenital	Acquired	Congenital	Acquired
Secundum ASD		++	++			(+)	+		(+)
Superior sinus venosus defect		++	+				+		
AVSD/primum ASD		++	++	(+)		(+)		(+)	++
VSD		+	(+)	+	(+)[a]				+
Ebstein anomaly	+++	++	+	(+)	++[b]		++		
TOF		++	++	++	++		+		+
TGA									
Atrial switch		+++	+	++[c]	+++[b]		+++		+
Arterial switch		+		+[c]	(+)		(+)		
ccTGA	++	+	+	(+)	++[b]			+	++
Fontan operation									
Atriopulmonary connection		+++	++		+[b]		++		
Intracardiac lateral tunnel		++	+		+[b]		++		
Extracardiac conduit		+	+		+[b]		+		
Eisenmenger physiology Incompletely palliated CHD		++	++		++[d]				

Empty cells indicate that although not specifically indicated, arrhythmic events may occur (no symbol).

(+) = minimal risk + = mild risk ++ = moderate risk +++ = high risk

CPET

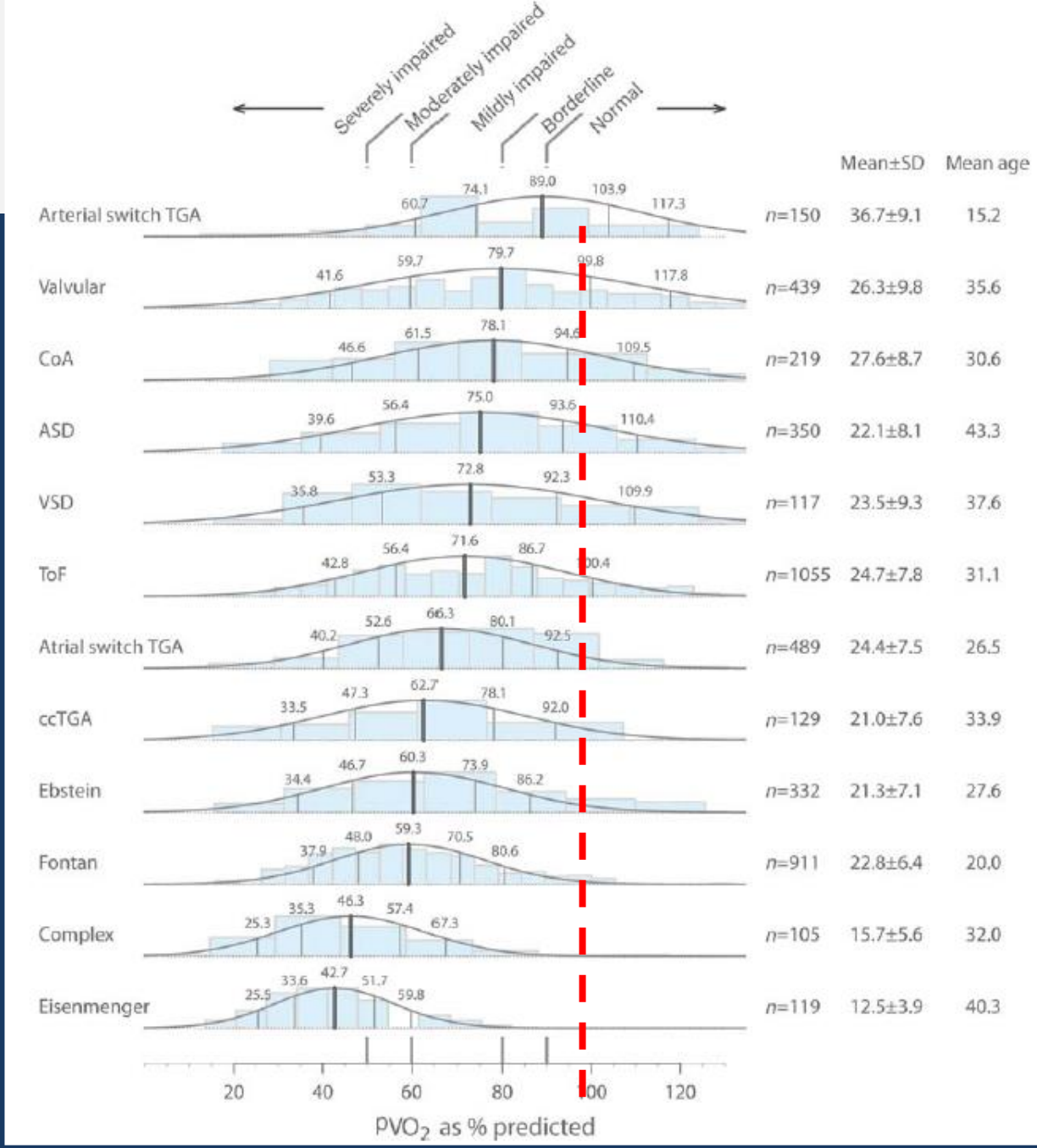
- In CHD, self-estimated physical capacity corresponds poorly with objective measures of exercise capacity.

- Therefore, cardiopulmonary exercise testing (CPET) has gained importance for objective assessment of exercise intolerance in both apparently asymptomatic and symptomatic CHD patients

- CPET can allow to identify also exercise-induced desaturation that might not be apparent at rest (predominant L-R shunt at rest with inversion of the shunt during exercise)

CPET

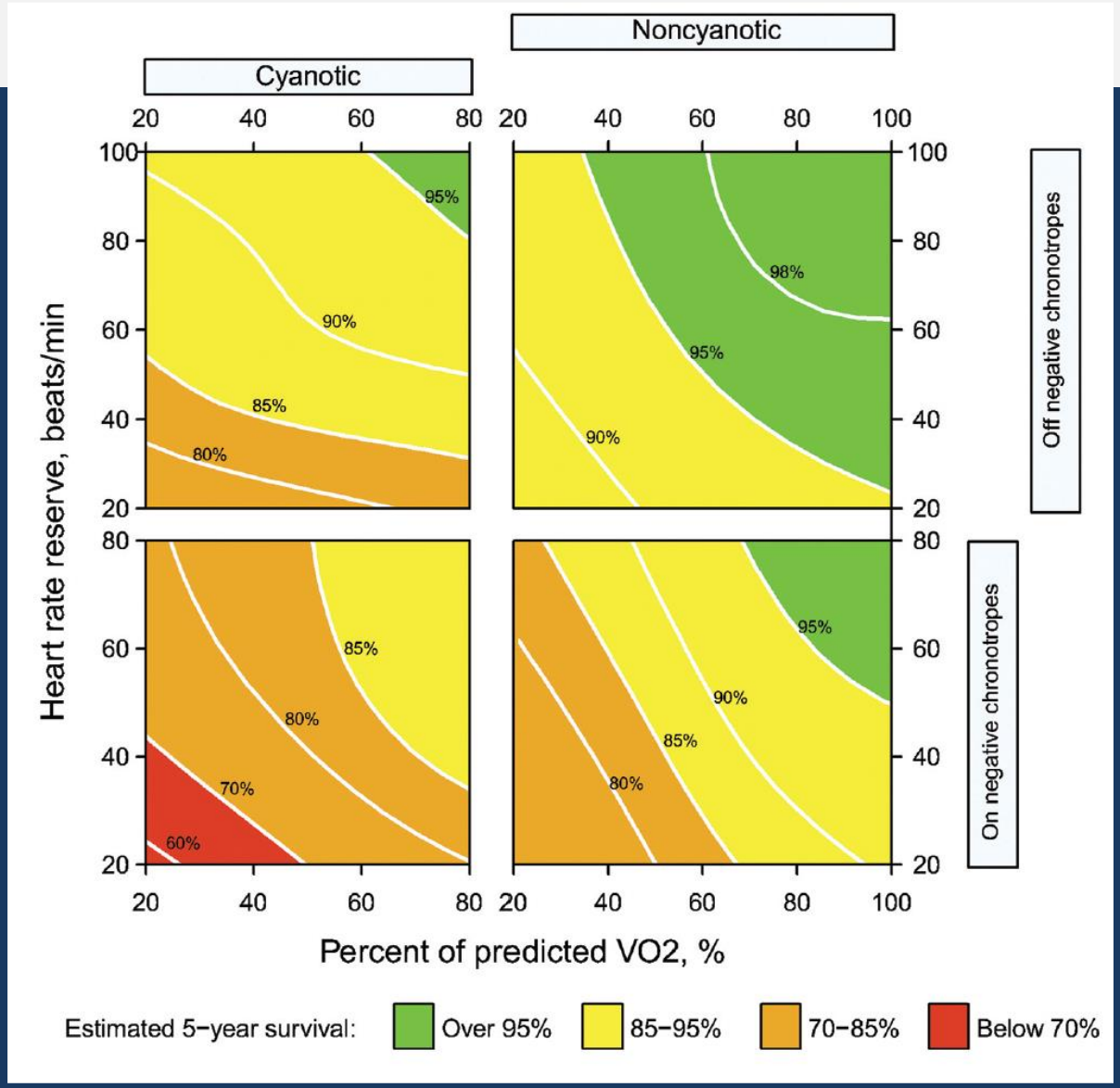
- VO2 retains strong prognostic capability in adults with CHD
- It has been suggested that %predicted VO2 could be further normalized for diagnosis-specific reference values



Kempny A et al. Eur Heart J 2012;33:1386-96

CPET

- VO2 retains strong prognostic capability in adults with CHD
- It has been suggested that %predicted VO2 could be further normalized for diagnosis-specific reference values
- An integrated combination of parameters, rather than a single parameter may provide more straightforward information



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