



Double discordance

« Congenitally corrected transposition of the great arteries »

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Maladies Cardiaques Héréditaires- CARDIOGEN



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CRMR Malformations Cardiaques Congénitales Complexes

CRMR Maladies Cardiaques Héréditaires

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INSERM Embryology & Genetics of Congenital Malformations





M3C

ACCUEIL

QUI SOMMES-NOUS ?

ACTUALITÉS M3C-
NECKER +

PROCÉDURES
INNOVANTES 2019

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ESSAIS & REGISTRES

ÉVÉNEMENTS

PLUS +



www.carpedemm3c.com

M3C

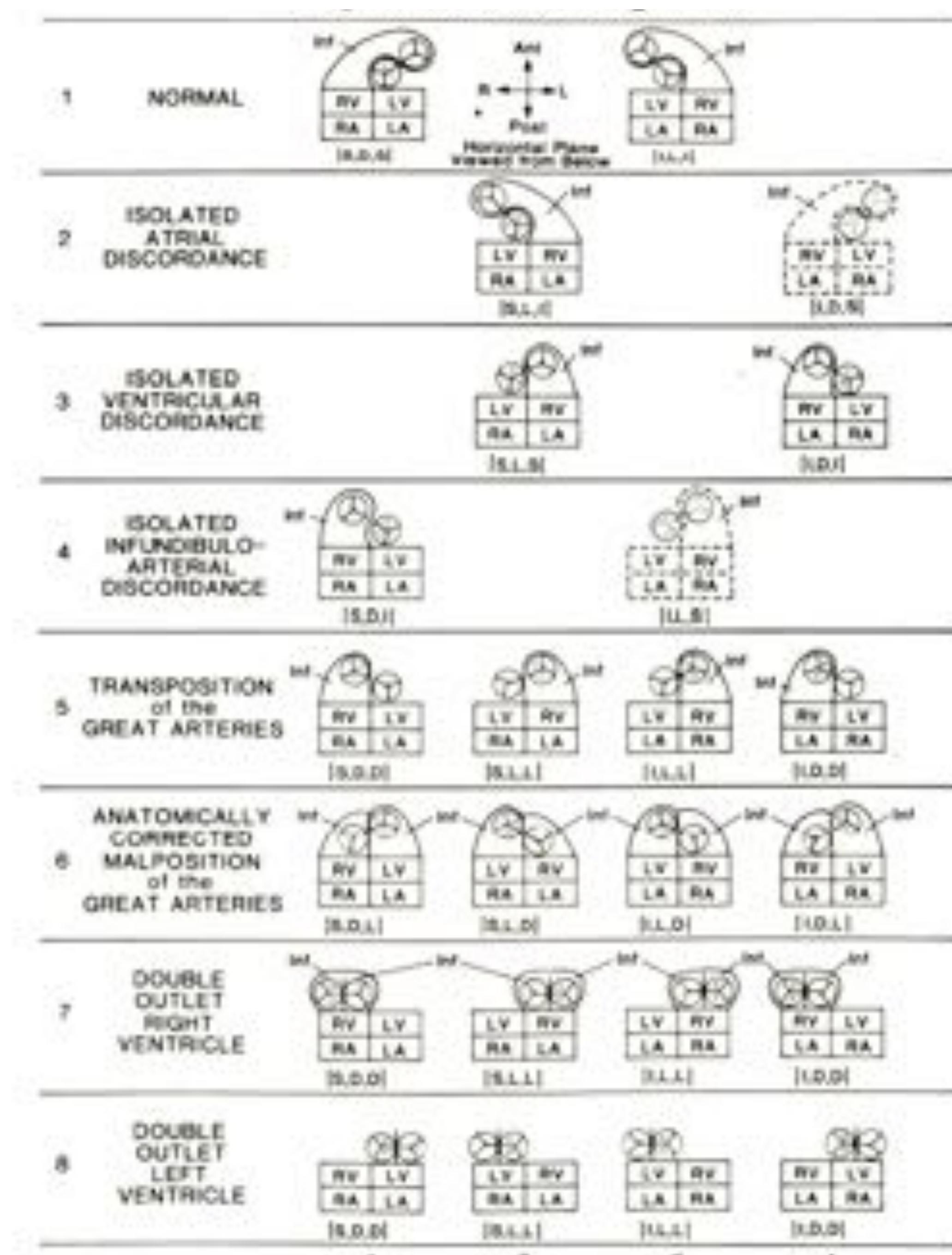
Centre de Référence

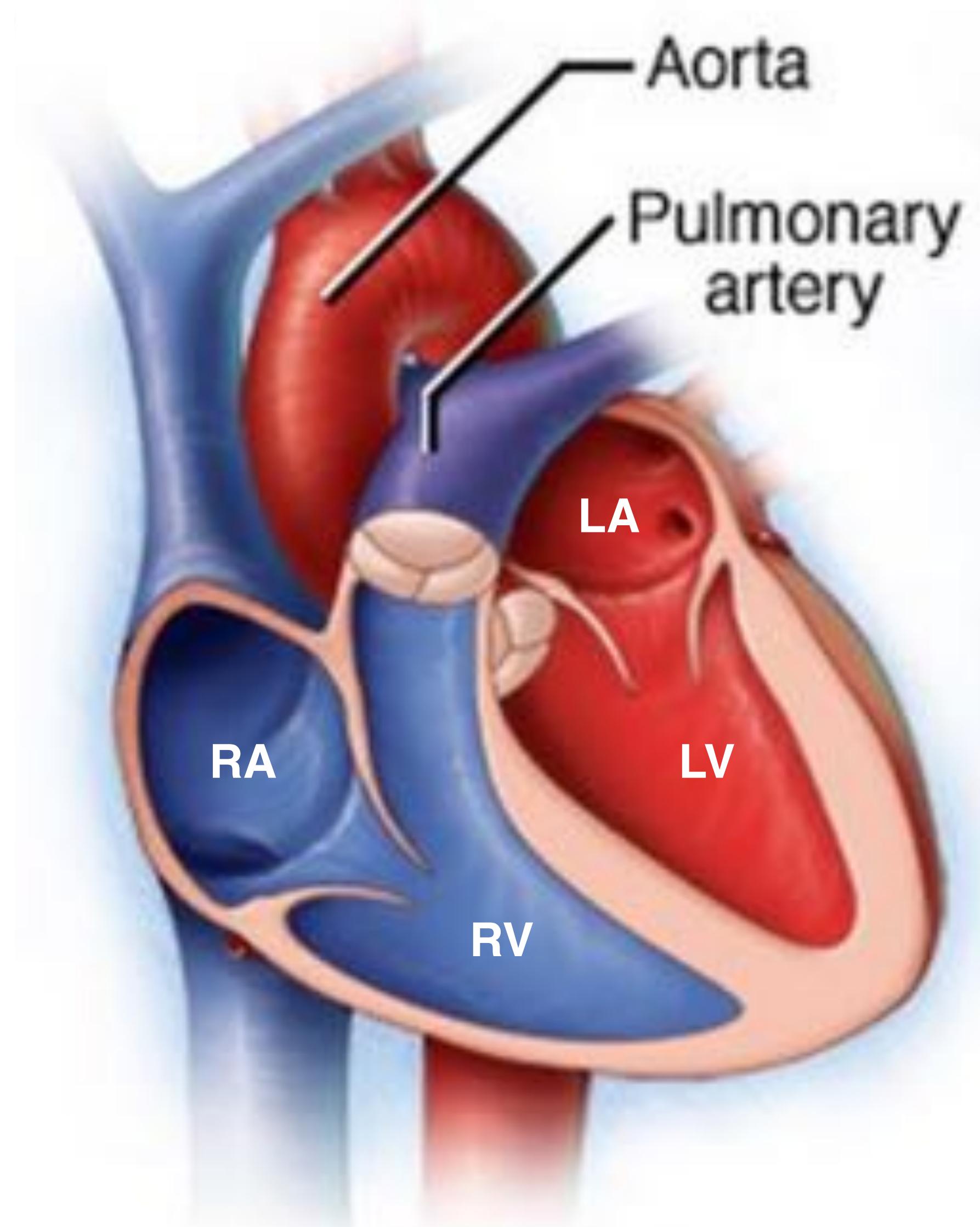
Malformations Cardiaques Congénitales Complexes

What is double discordance ?

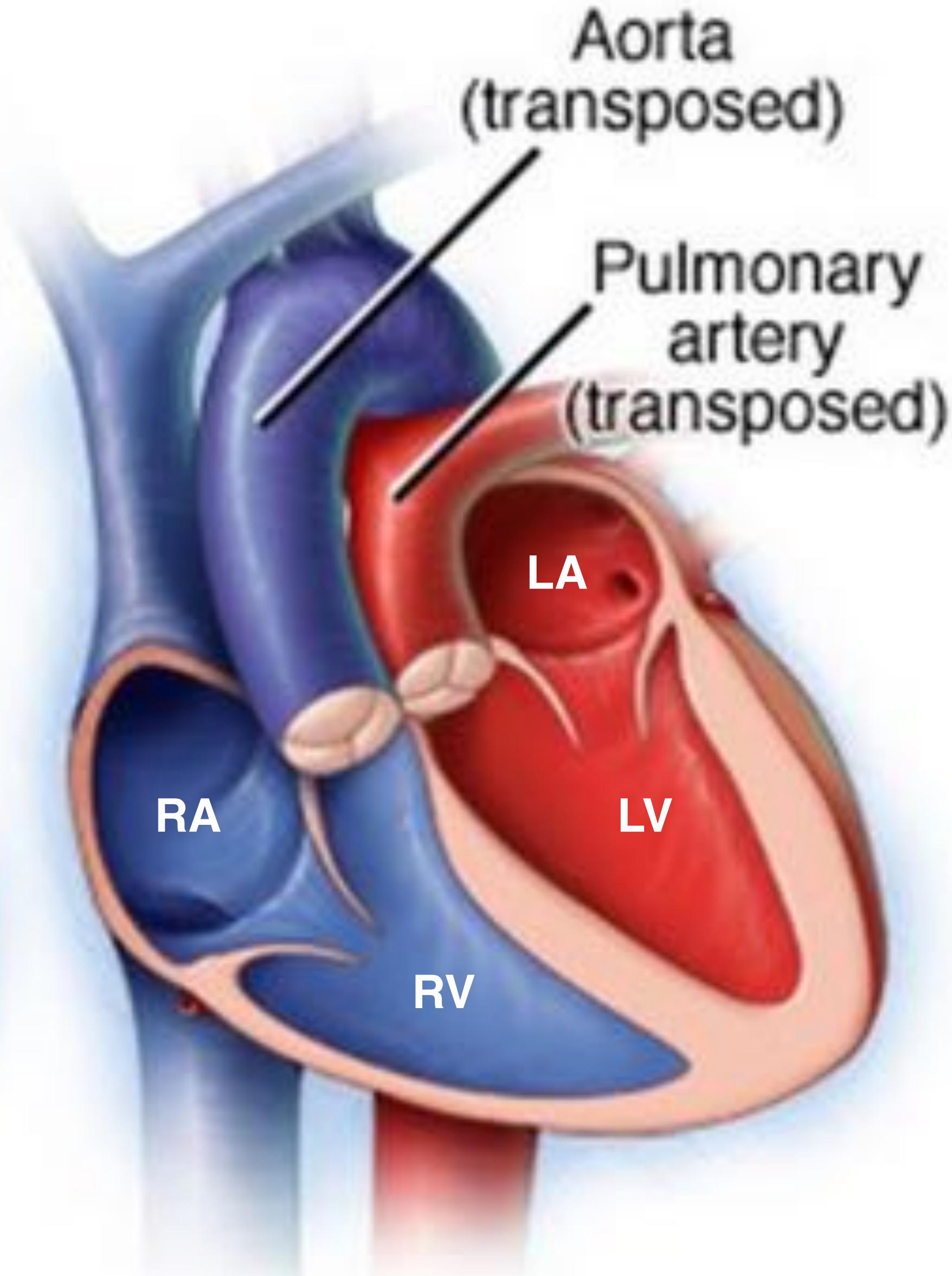
Types of human hearts

Segmental sets and alignments

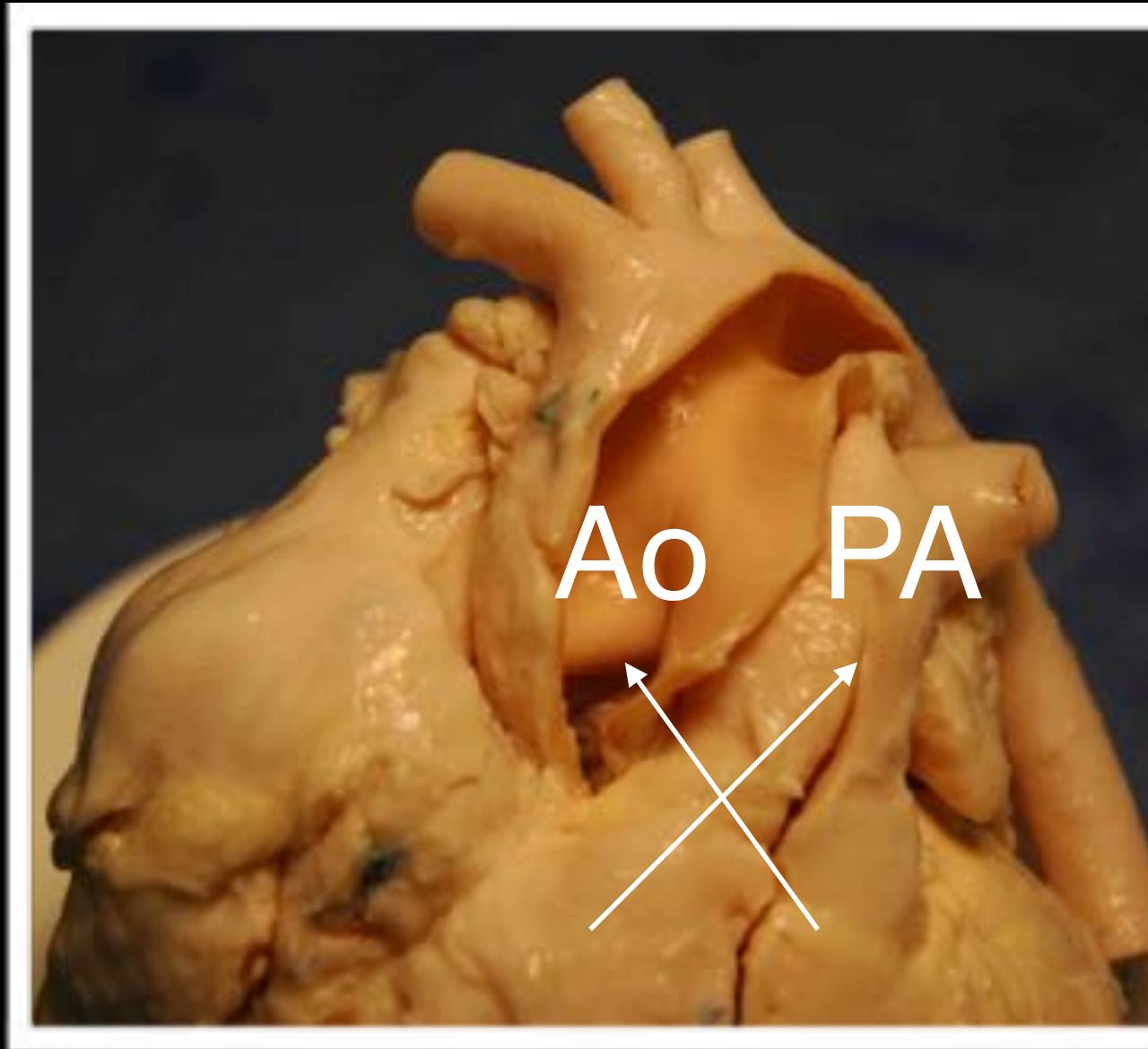




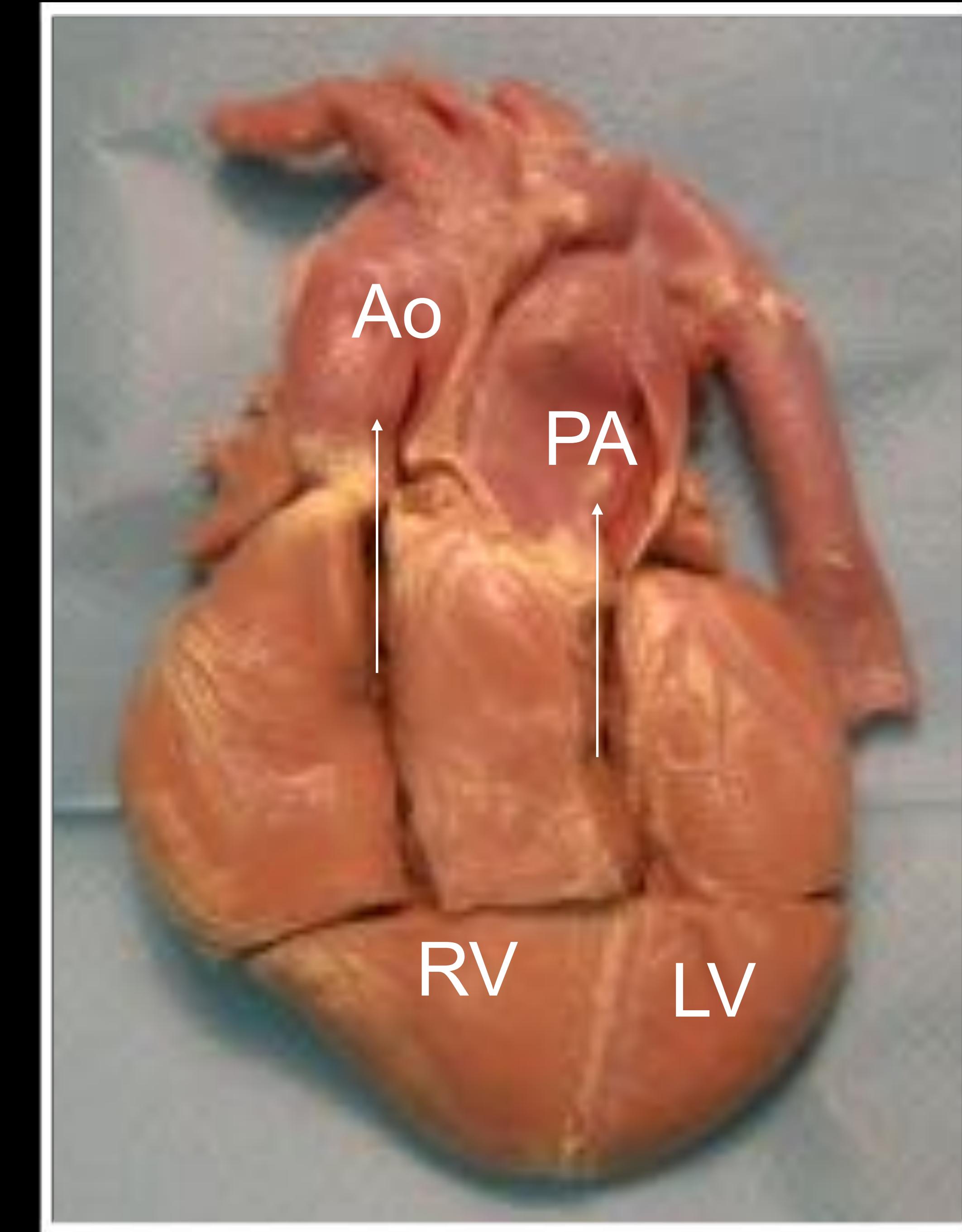
Normal heart



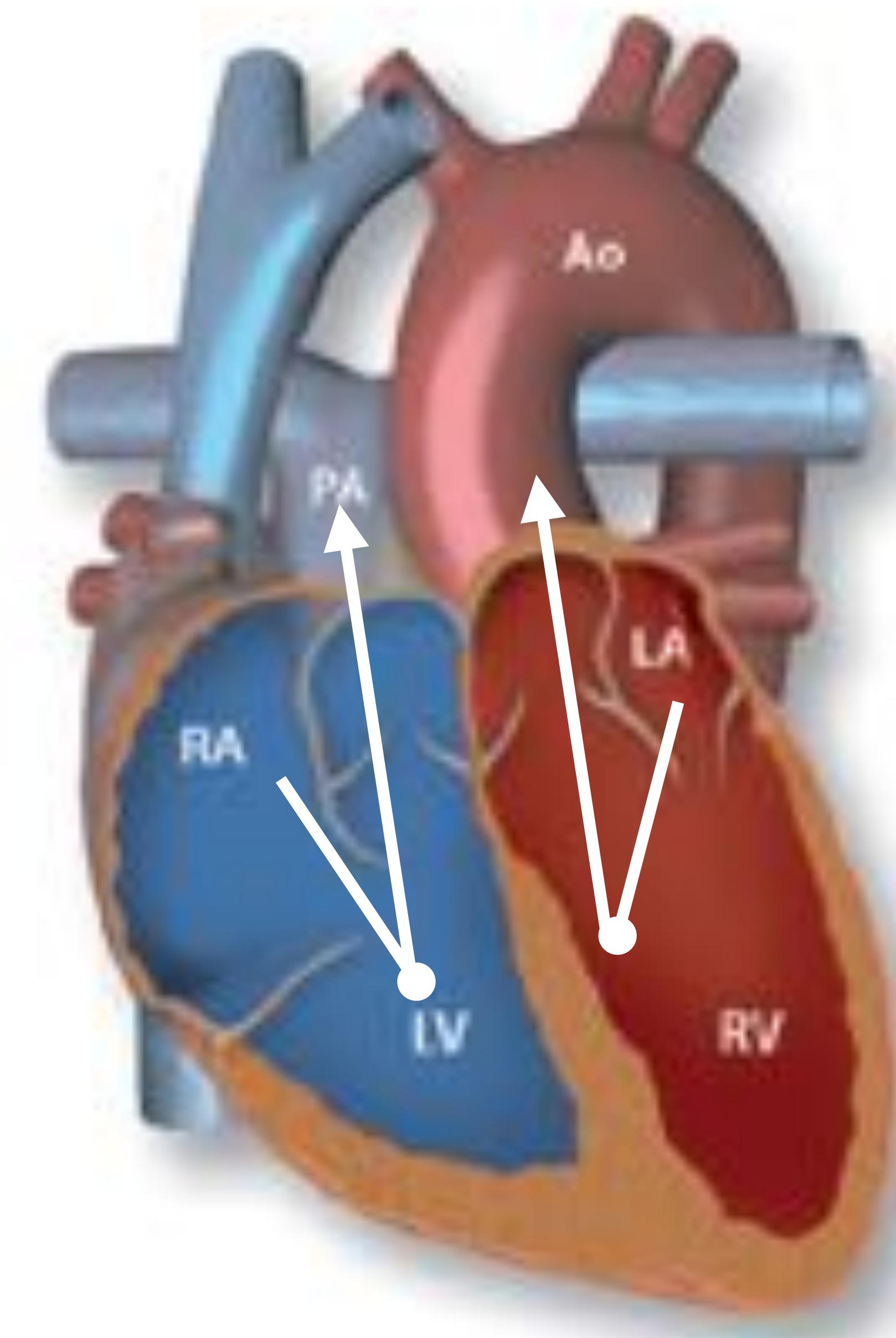
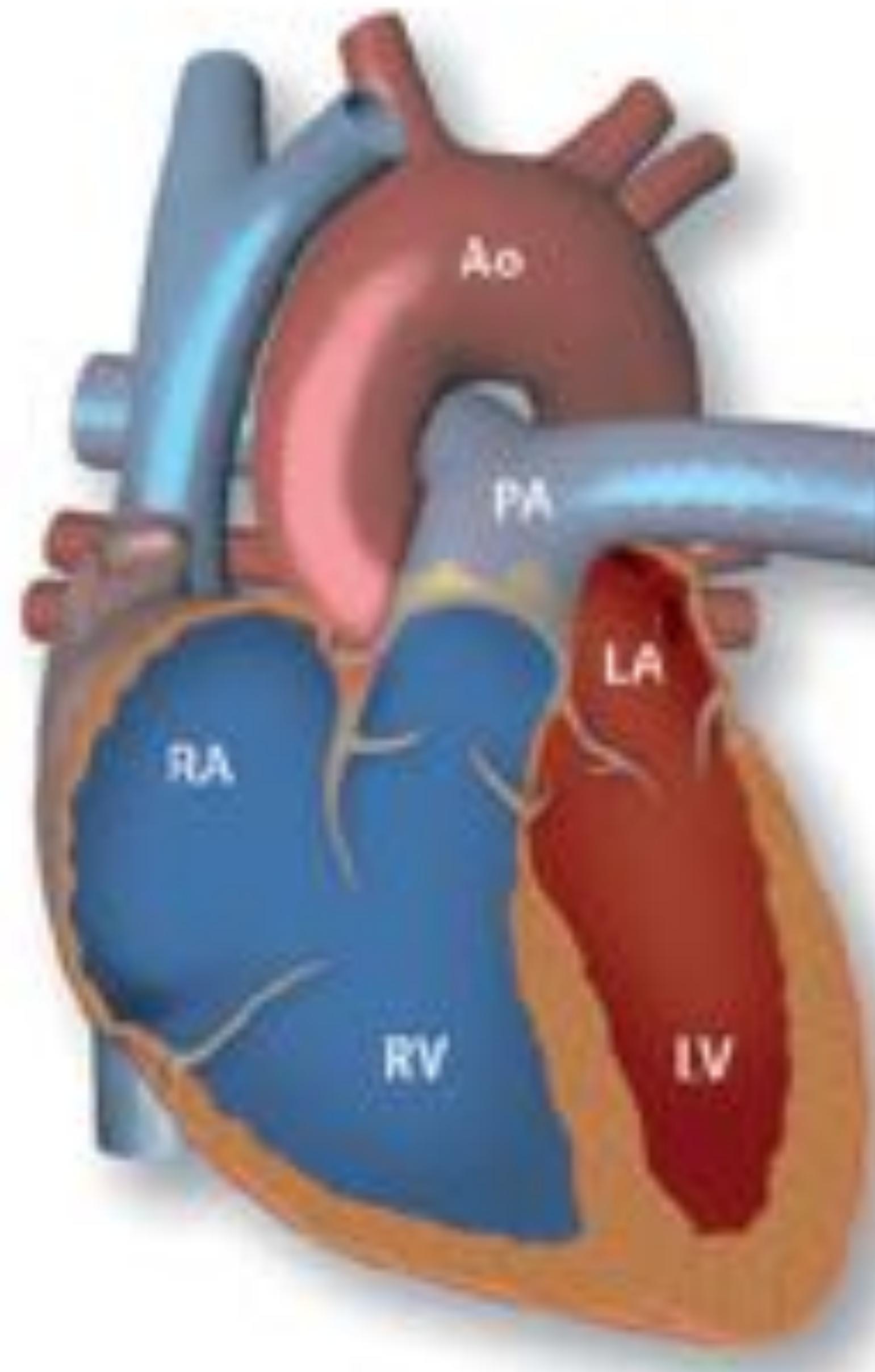
Transposition
of great arteries



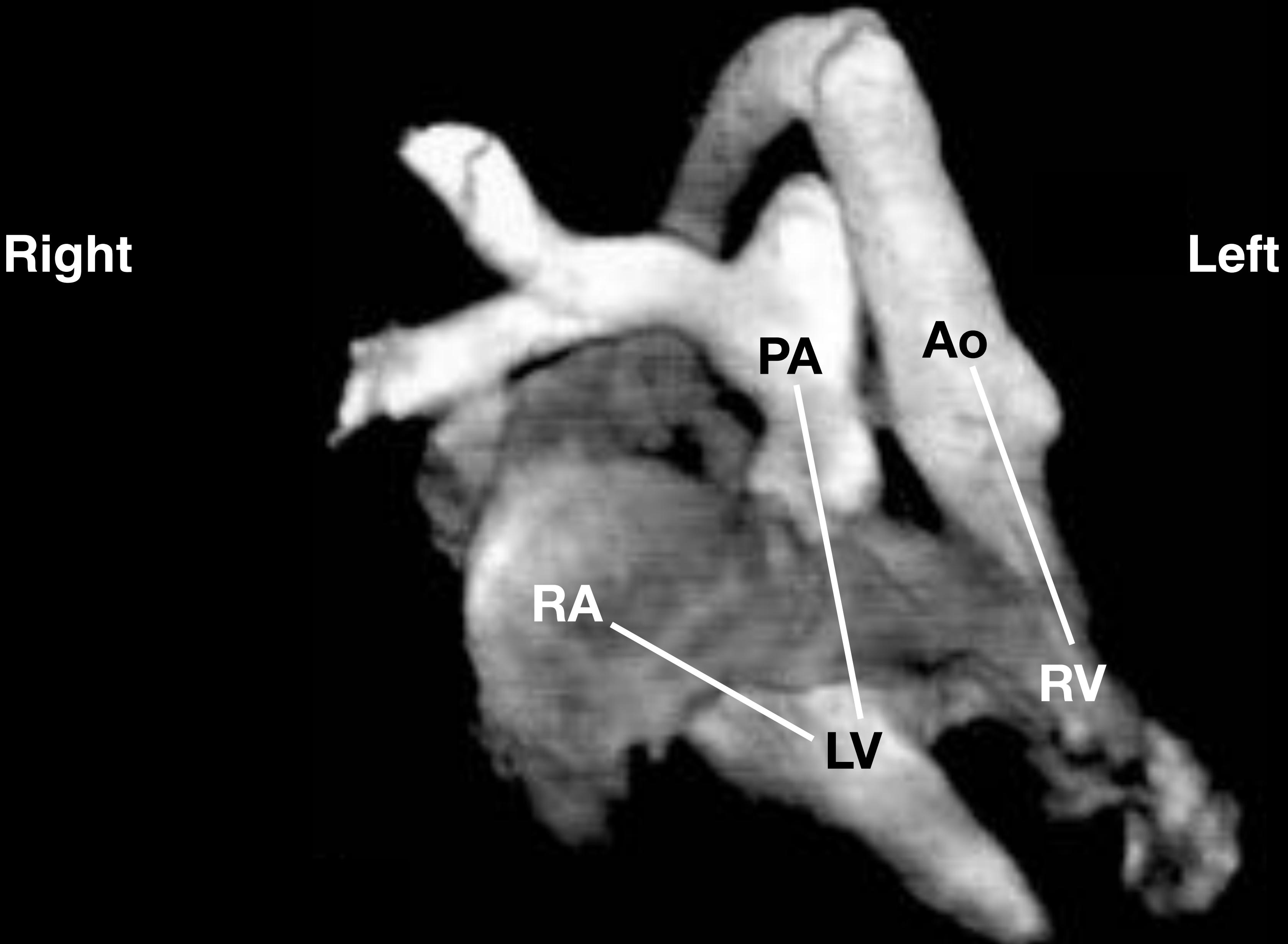
TGA (S,D,D)



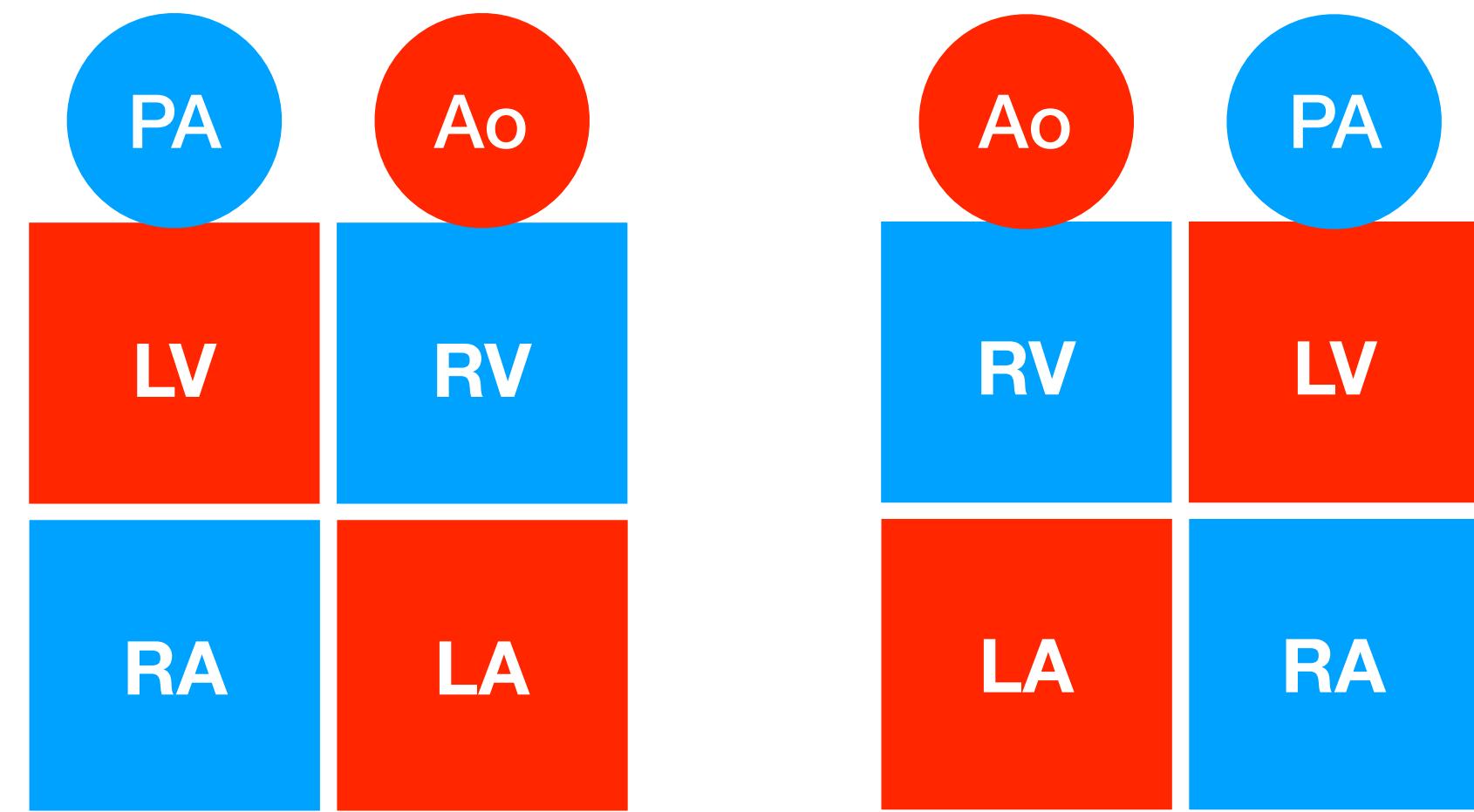
Double discordance



Double discordance

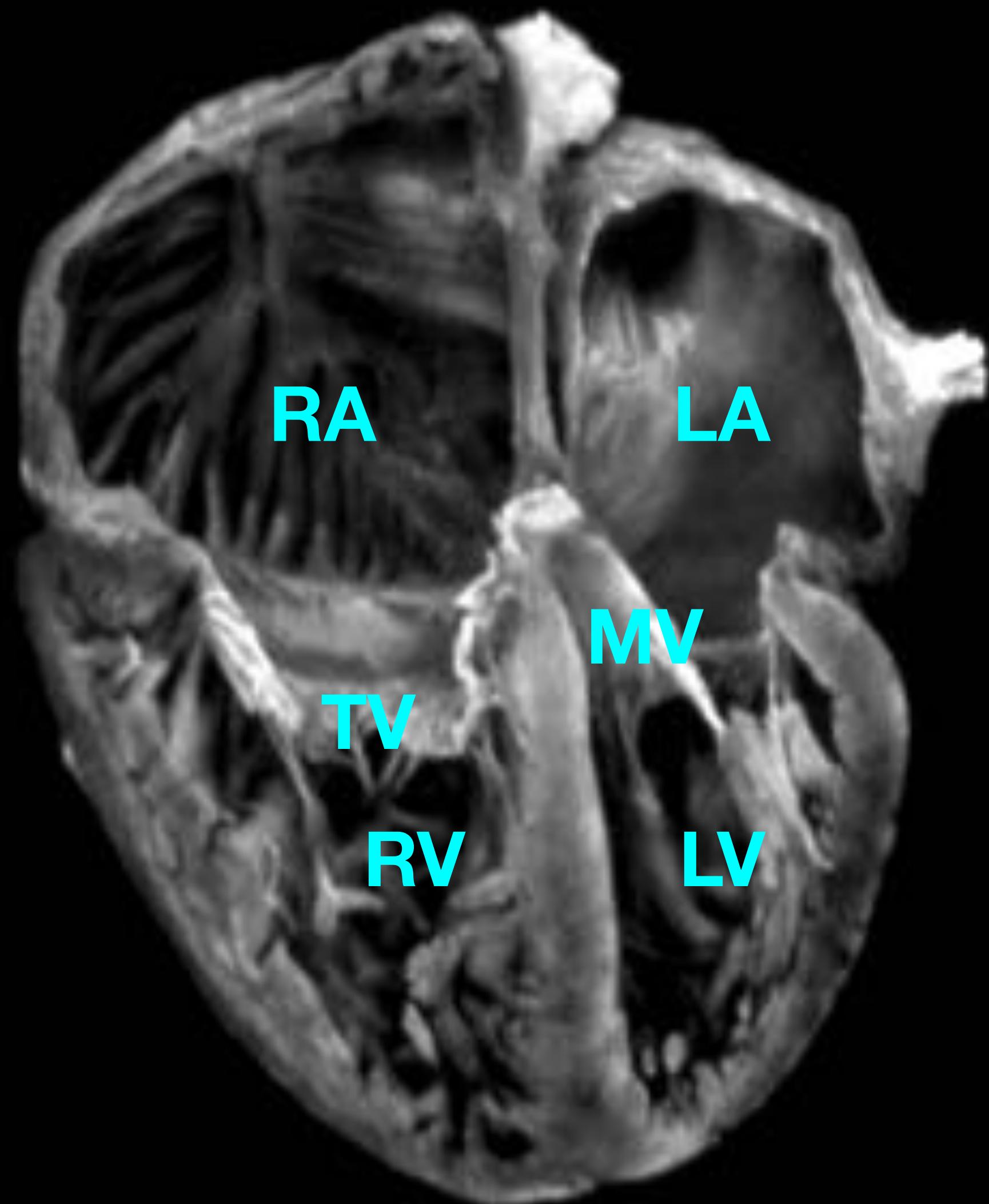


**Double discordance
or
Congenitally corrected transposition
of the great arteries**

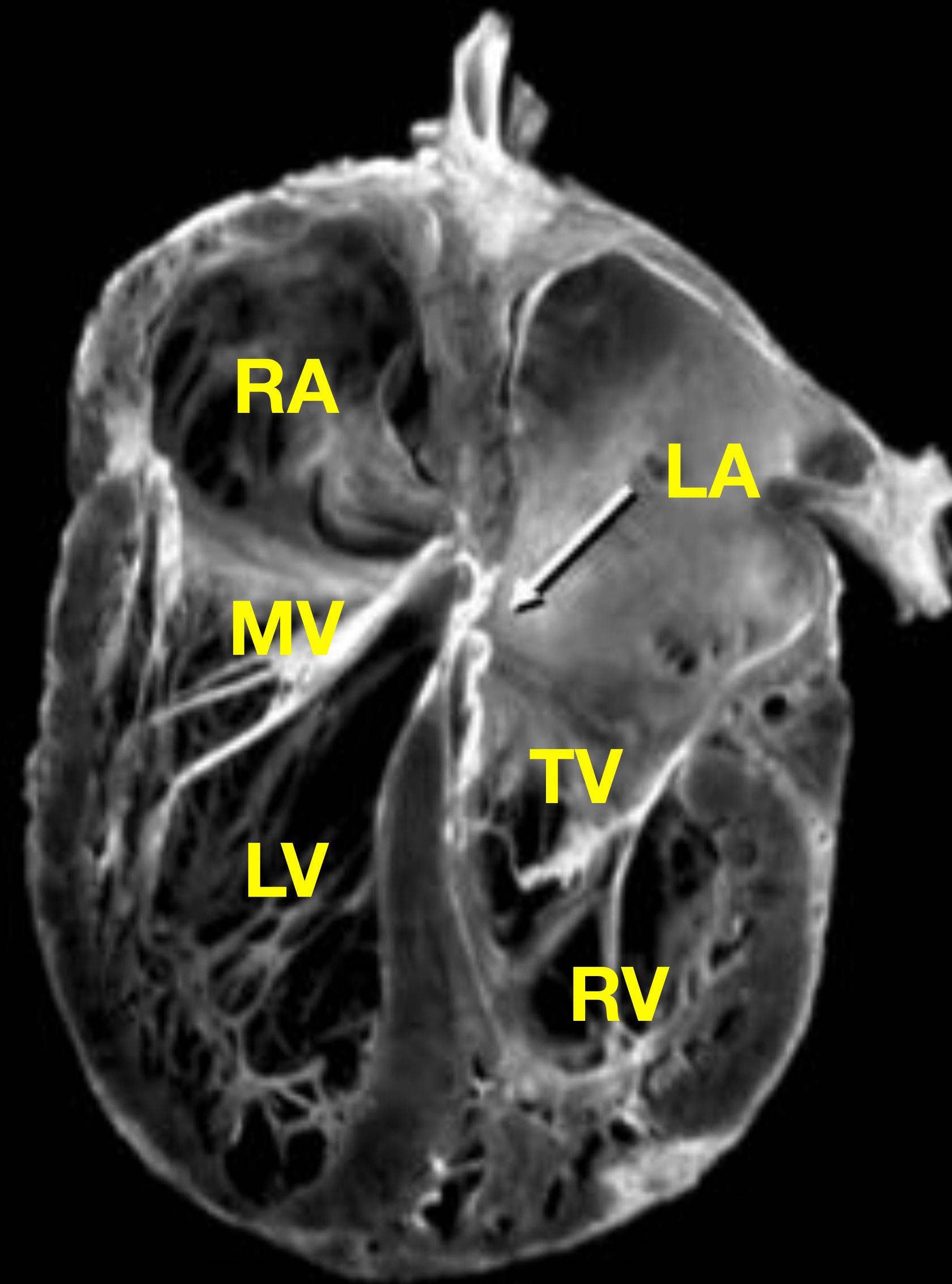


S,L,L

I,D,D

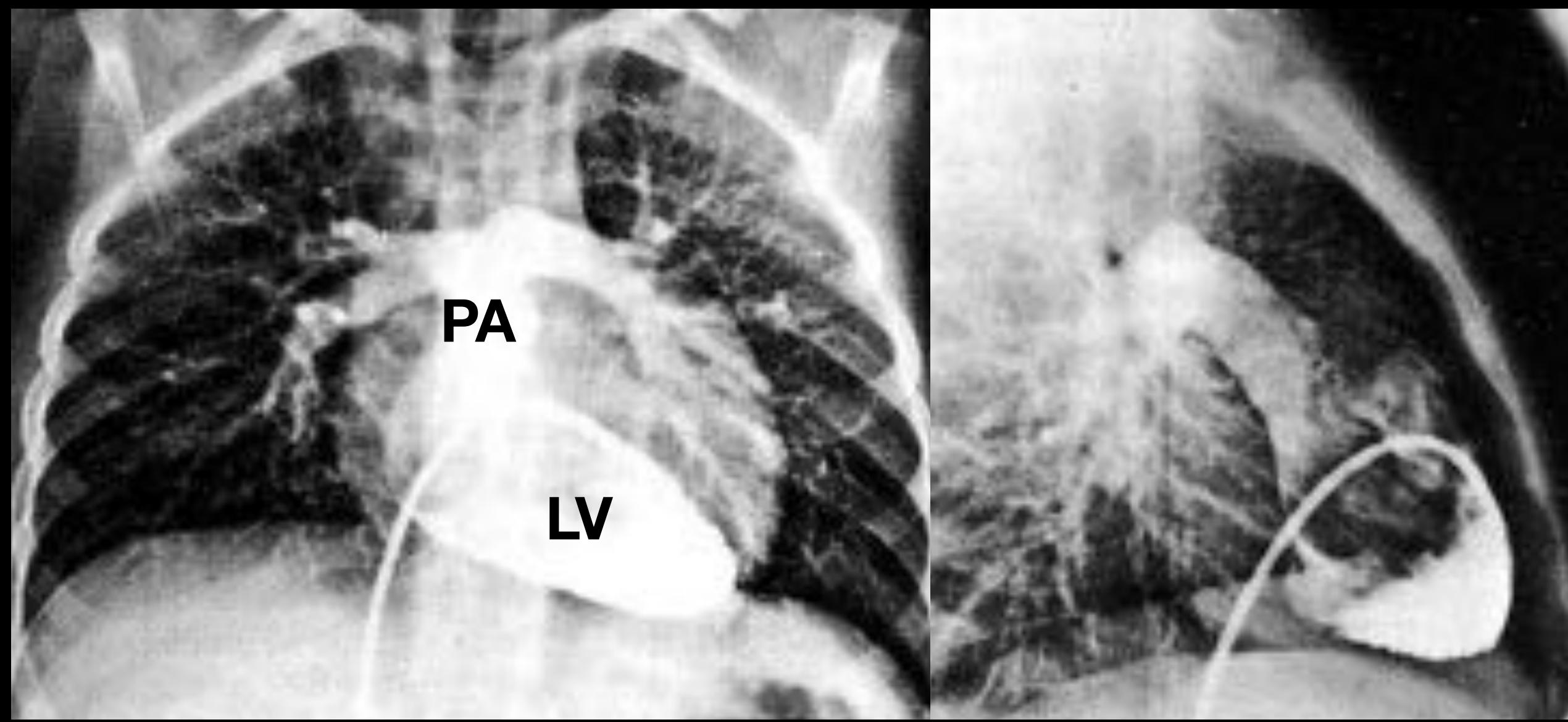
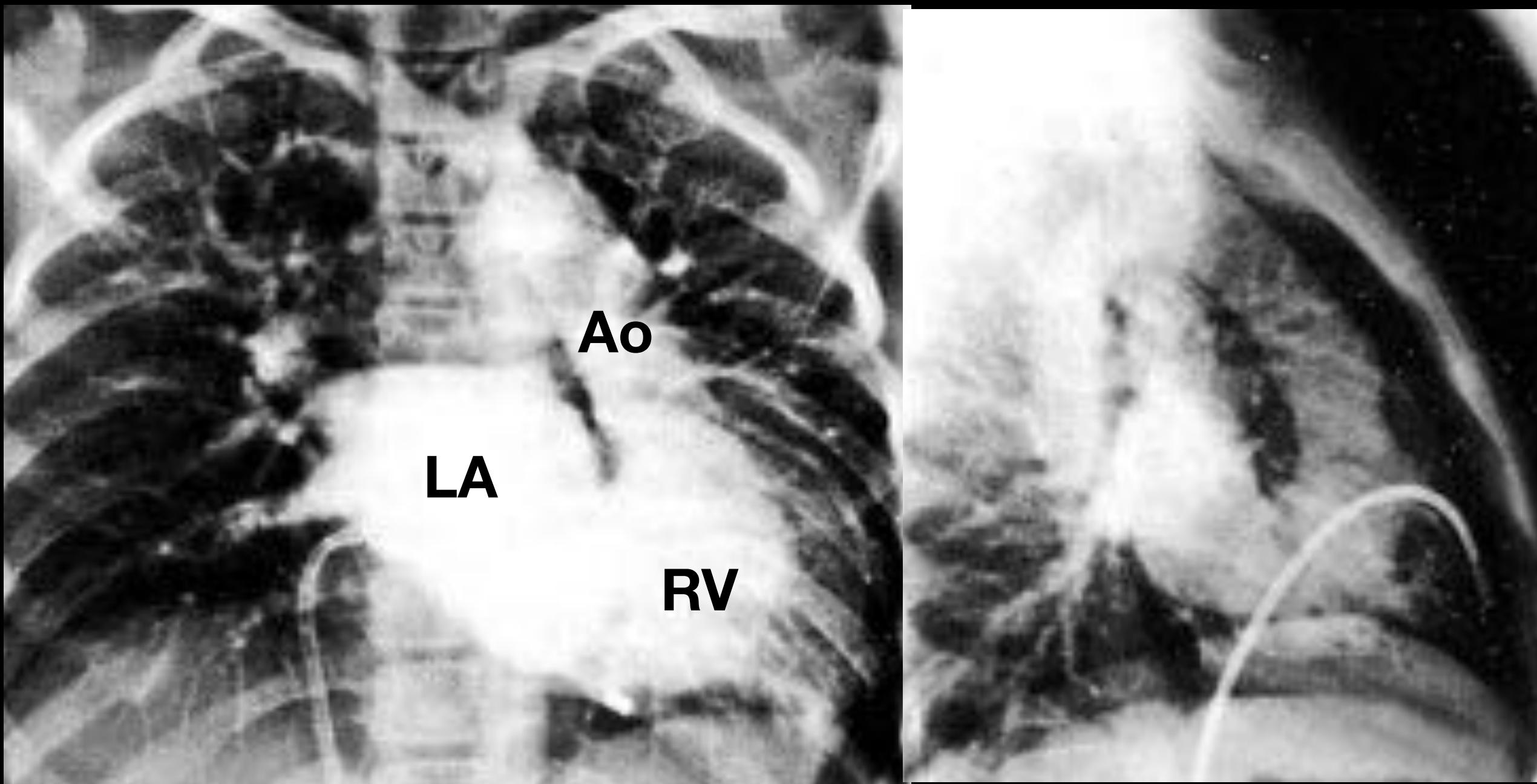


Normal heart



Double discordance

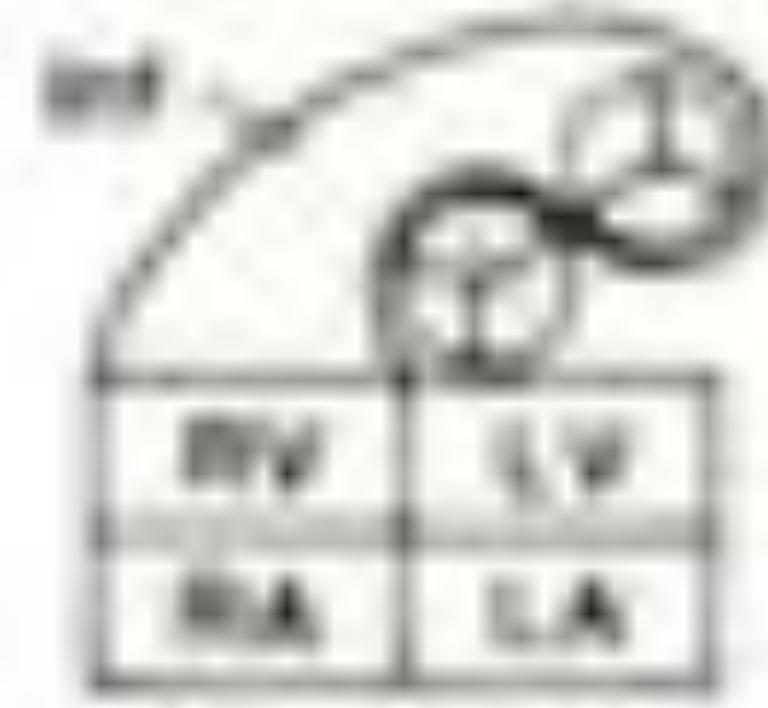
Double discordance



Types of human hearts

Segmental sets and alignments

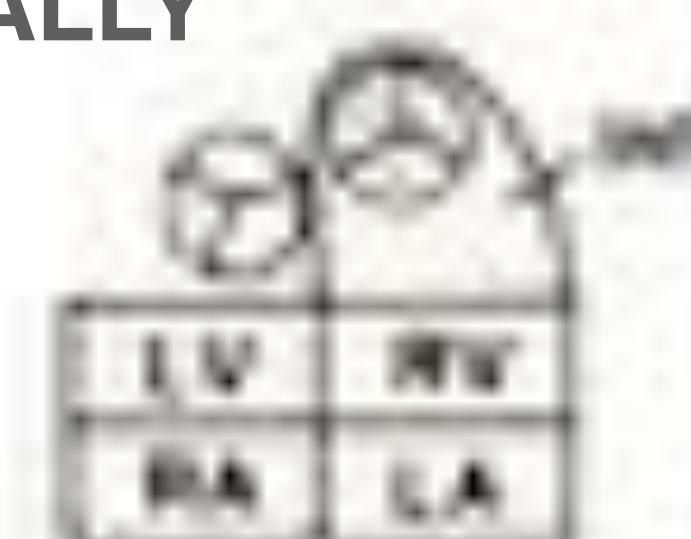
NORMAL



[S.D.A]

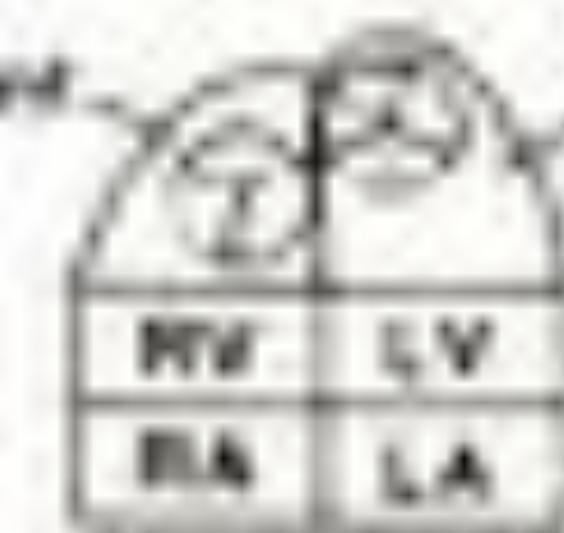
CONGENITALLY/PHYSIOLOGICALLY
CORRECTED

TRANSPOSITION
of the
GREAT ARTERIES



[S.A.A]

ANATOMICALLY
CORRECTED
MALPOSITION
of the
GREAT ARTERIES



[M.O.L]



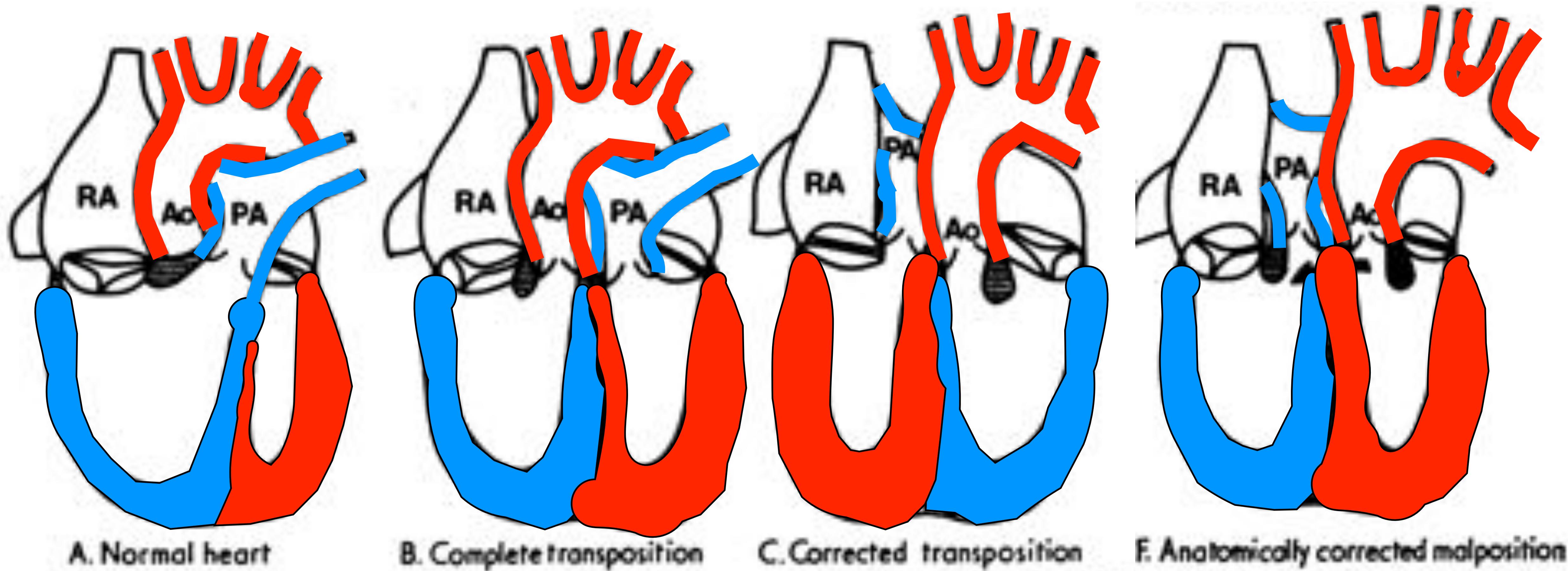
[M.L.O]

TRANSPOSITION
of the
GREAT ARTERIES



[S.O.P]

Malpositions and double discordance



Double discordance - definition

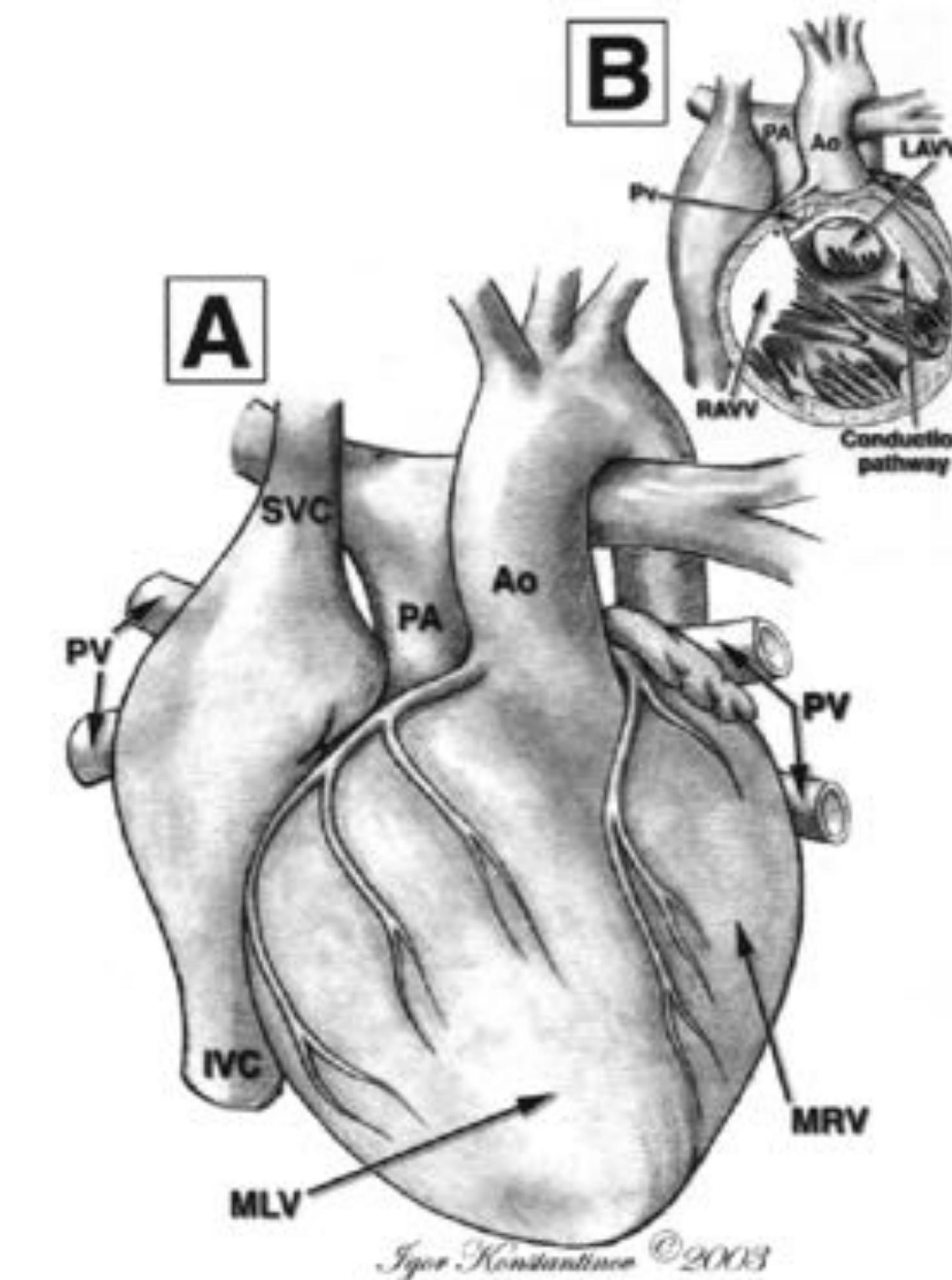
- Transposition : VA discordance
- Ventricular inversion (loop)

AV discordance

Double discordance

physiologically « corrected »

- But : $- \times - \neq + !!$



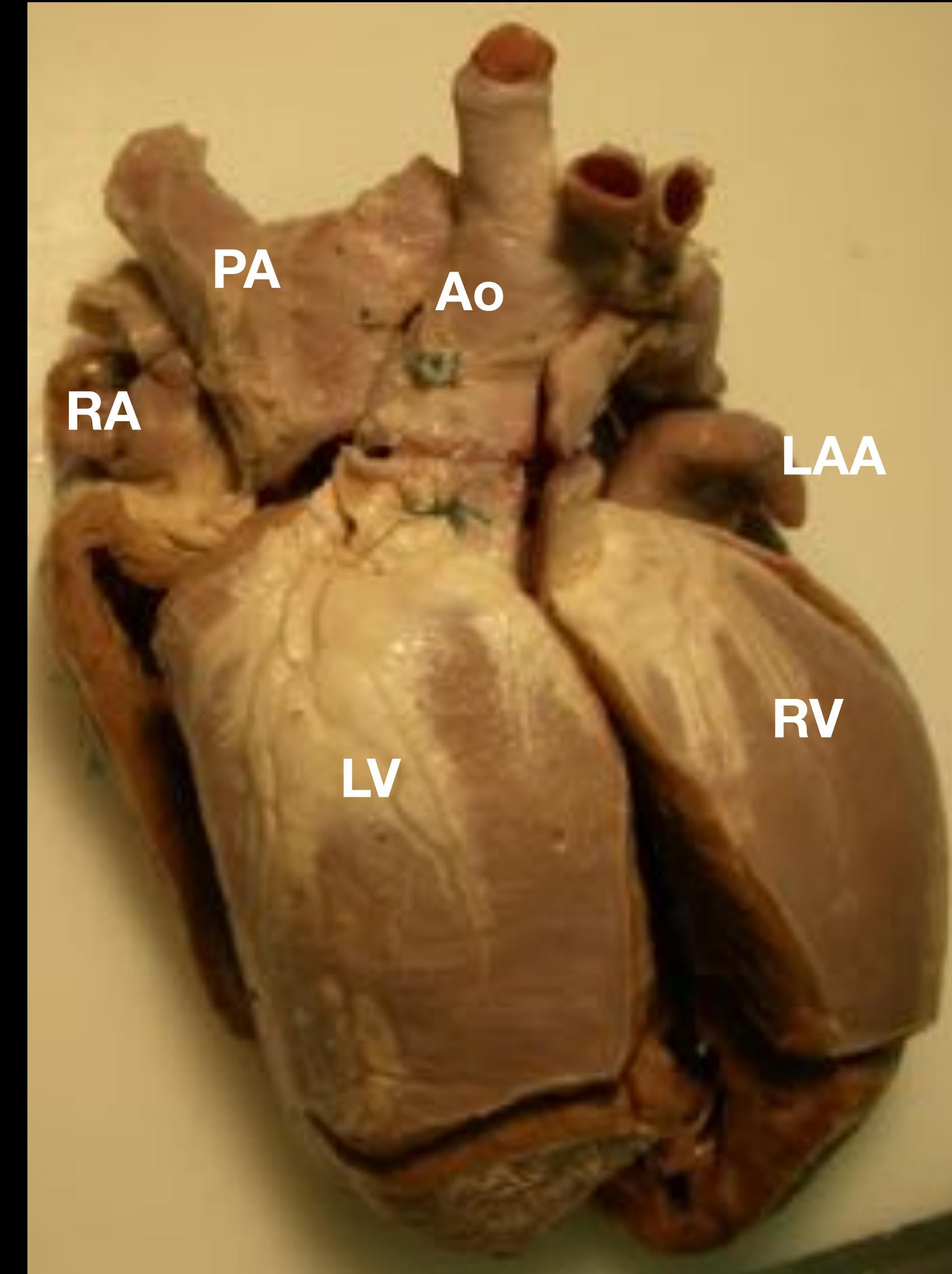
Double discordance

Anatomy

Courtesy Lucile Houyel M3C-Necker

Double discordance - Segmental analysis

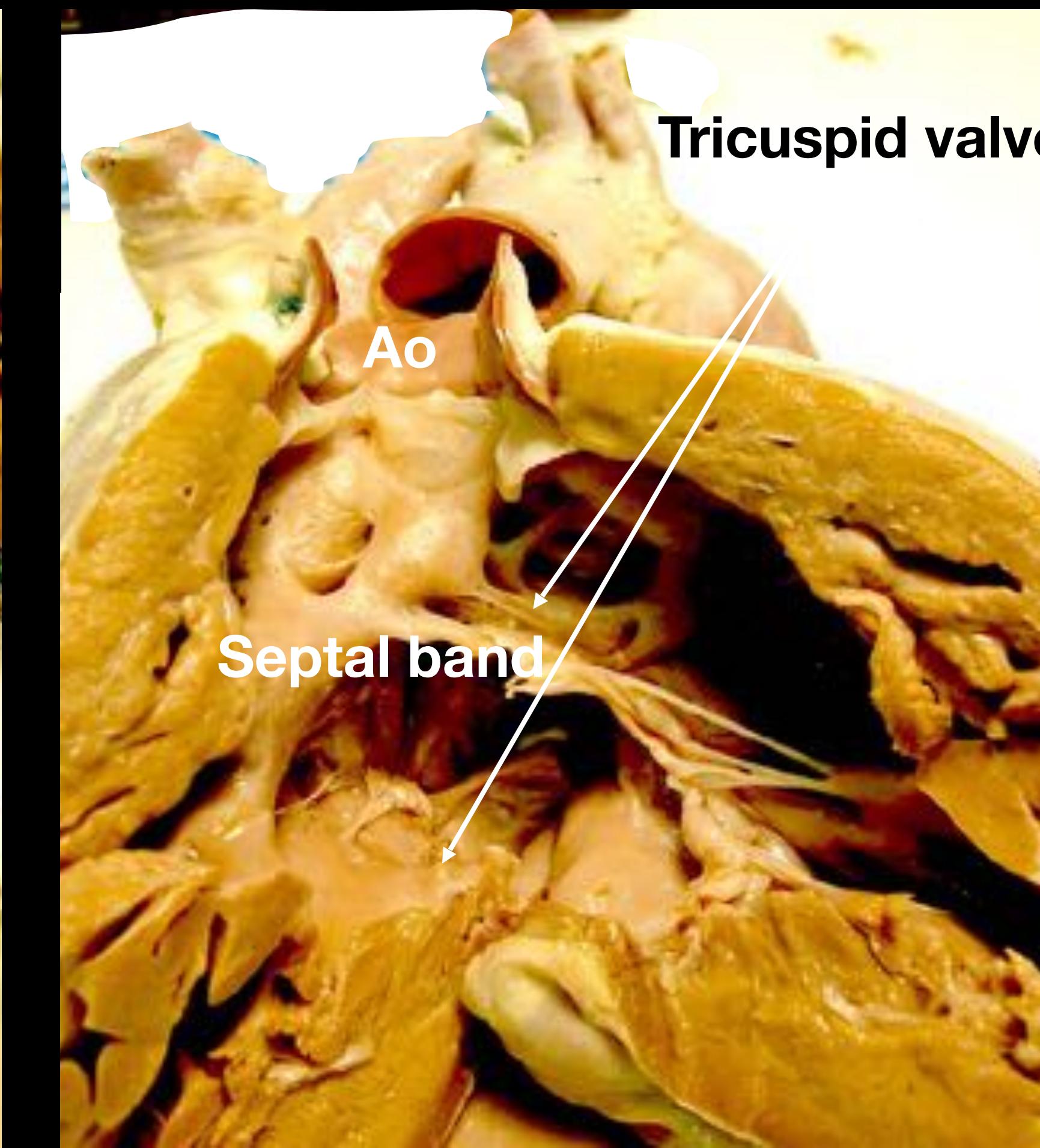
- Most frequently (97%) : {S,L,L}
- In situus inversus : {I,D,D}
- Rarely : {S,L,D}



Double discordance {S,L,L} - simple/isolated

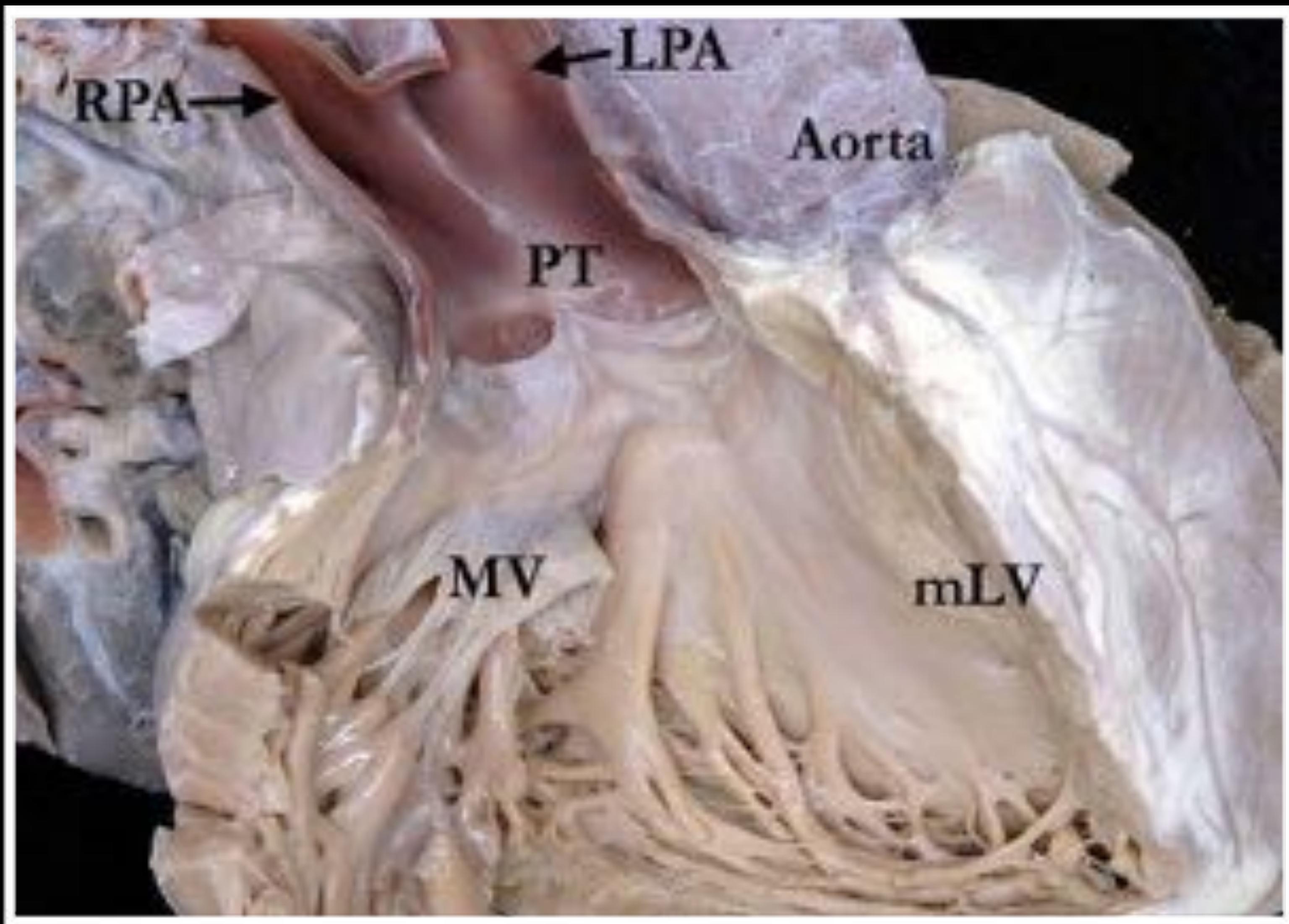


Right-handed LV
Fibrous continuity between
mitral and pulmonary valve

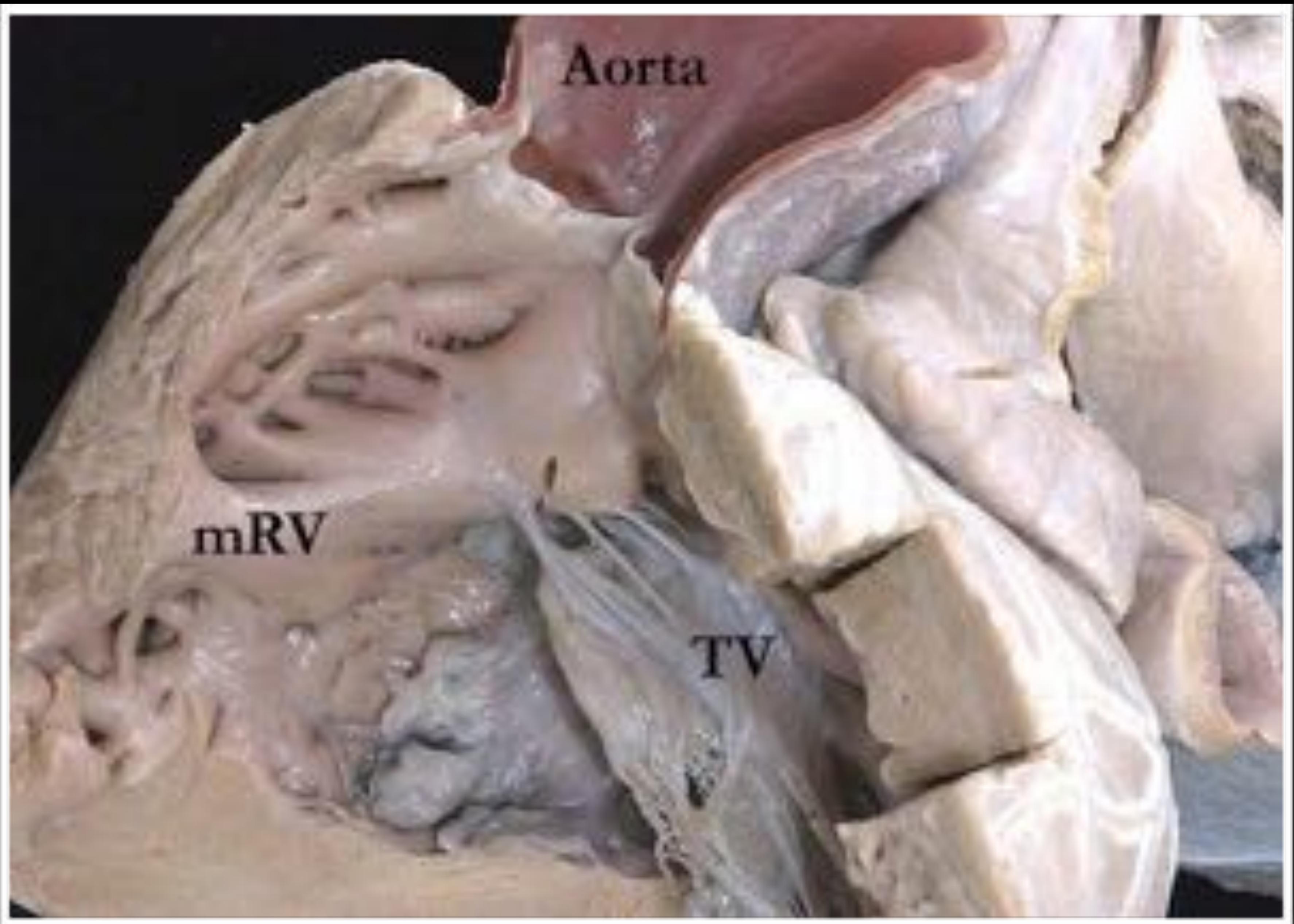


Left-handed RV with sub aortic conus

Double discordance



Double discordance



Double discordance

**Discordant Atrioventricular
Connections 1**

Double discordance

Embryology

- Laterality defect
- Early specification and looping process
- Families with siblings or first degree relatives with TGA
- Genes of heterotaxy probably involved : ZIC3, IIb activin receptor, CFC1 (cryptic), Pitx2

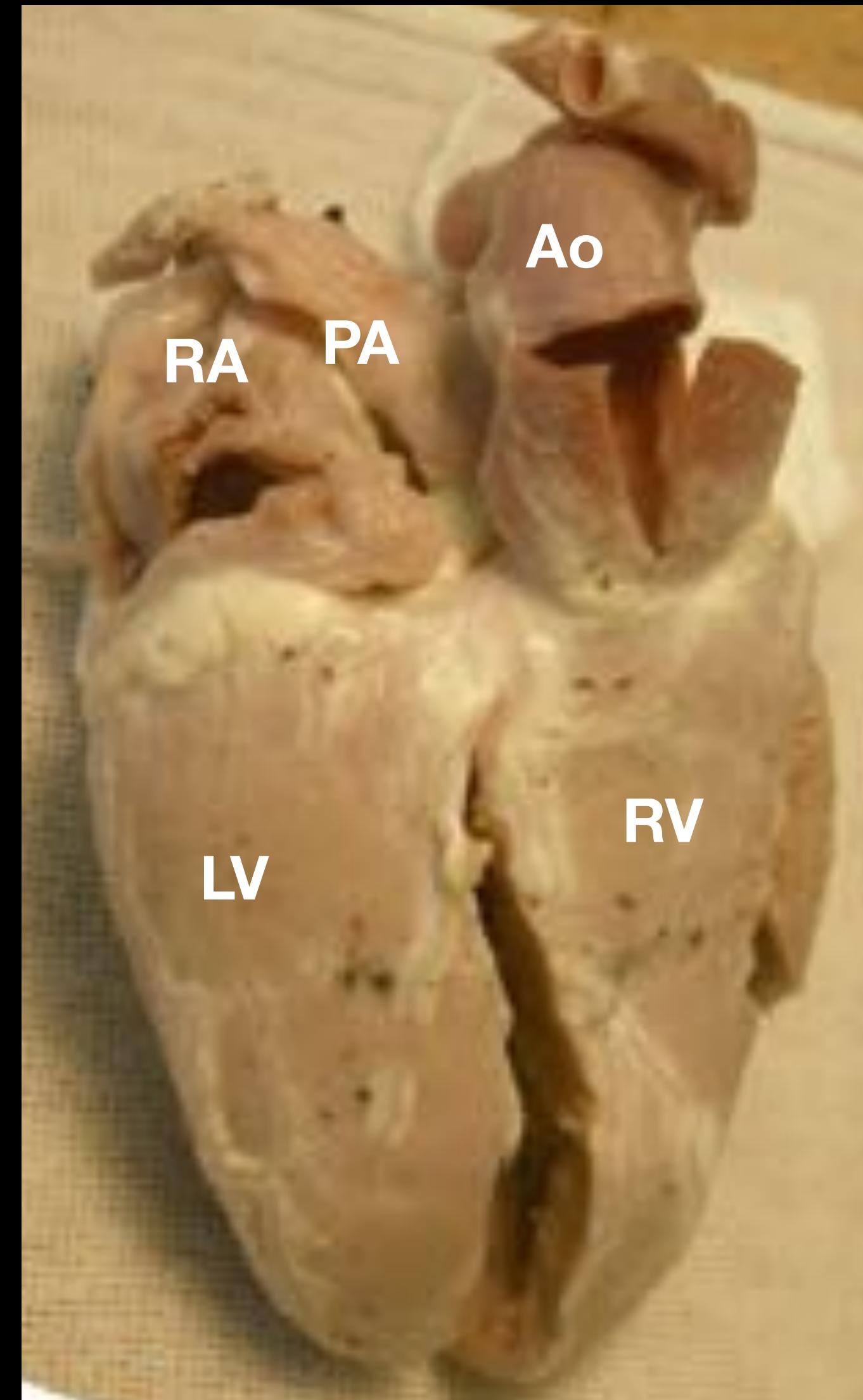
see the Chapters Heterotaxy and Genetics of CHD on www.carpedem3c.com

Double discordance is not only ventricular inversion

- Profound modification of the ventricular architectures
- Usual anatomical landmarks of the right ventricle are not present or are modified (Y of the septal band, membranous septum, His bundle)
- AV discordance causes malalignment of Inter-atrial and Inter-ventricular septa

Double discordance

- Cardiac architecture is profoundly modified
- Position of the heart M3C series
 - Mesocardia = 11/20 (55%)
 - Dextrocardia = 5/20 (25%)
 - Lavocardie = 4/20 (20%)

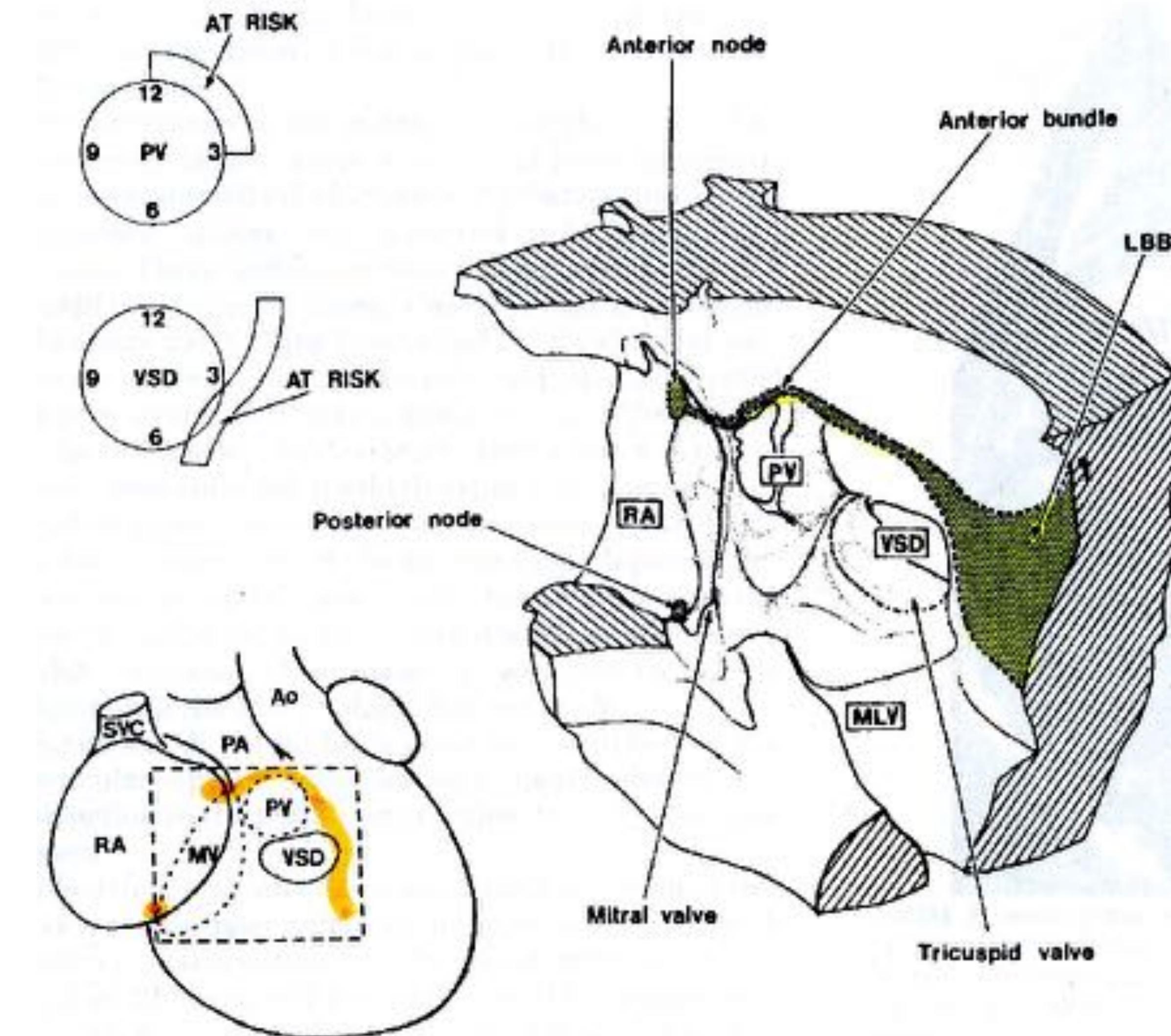


Associated anomalies

- Ventricular septal defect 75-80%
- Pulmonary outflow tract obstruction 40%
- Tricuspid valve malformation 70%
- Mitral valve malformation 20%
- Abnormal position of the conduction system in 100%

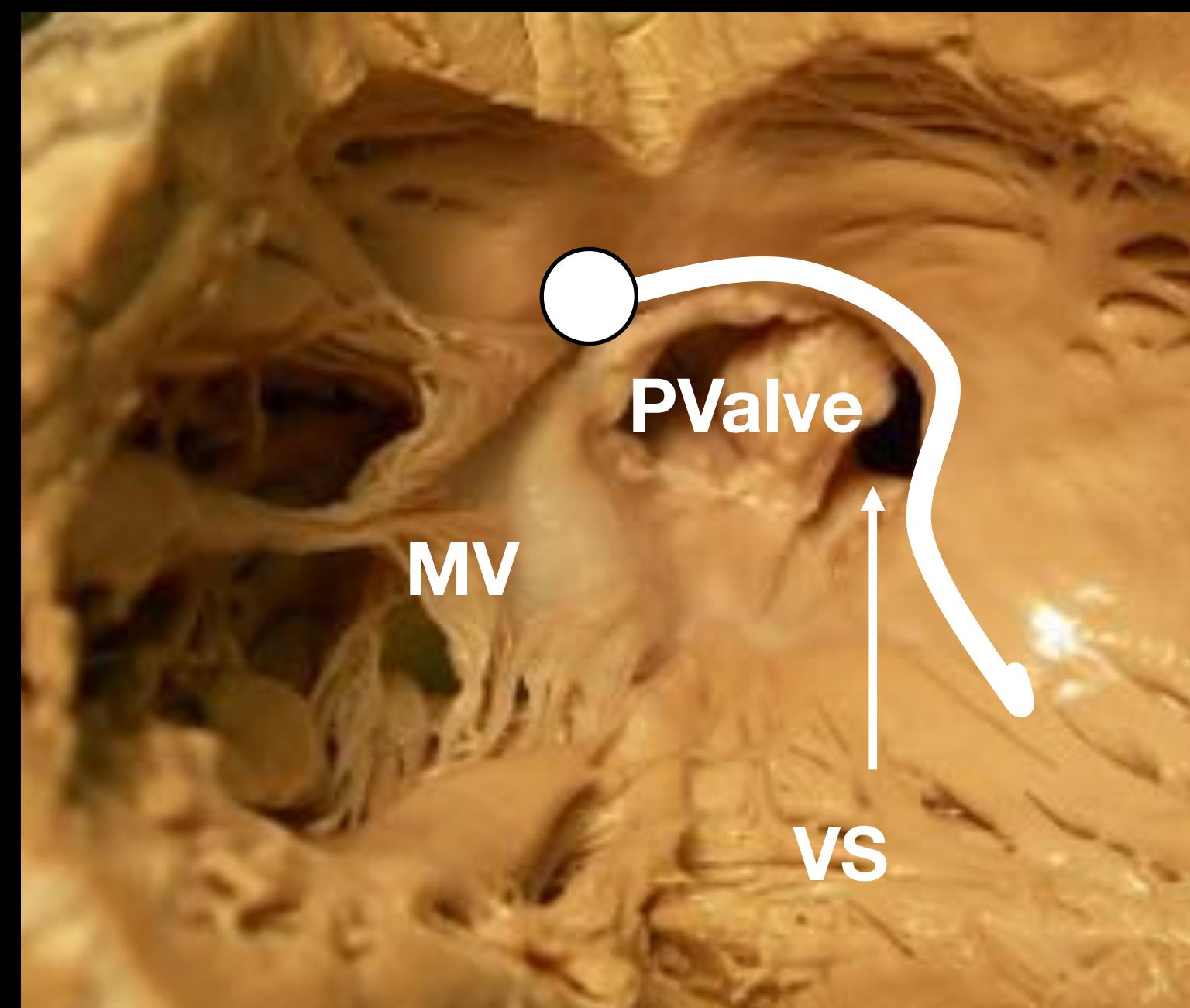
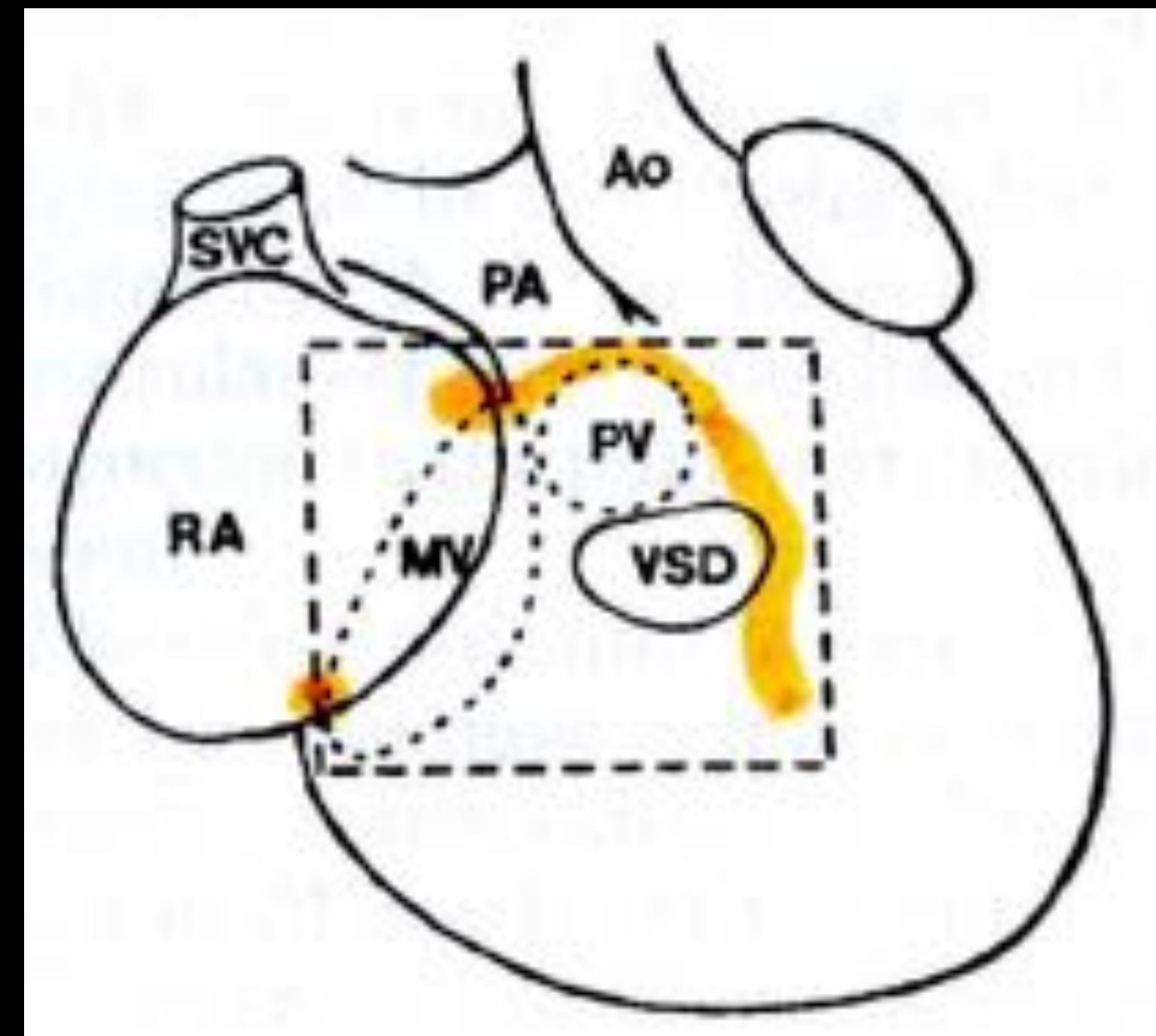
Double discordance Conduction system

- AV discordance
- ↓
- Malalignment of IA and IV septa
- ↓
- Malposition of the membranous septum
- ↓
- The posterior AV node cannot connect to ventricles
- ↓
- Second AV node anteriorly positioned

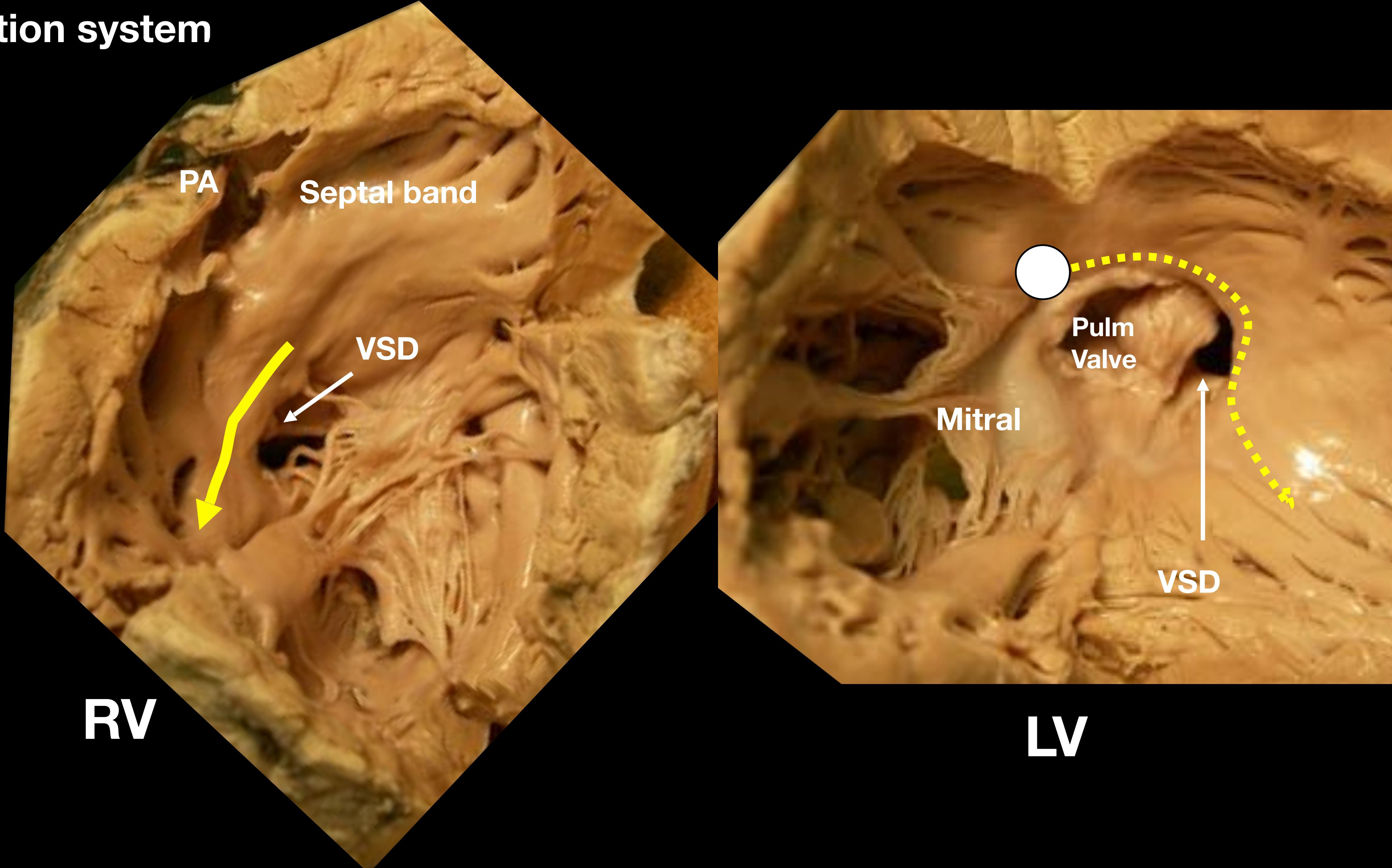


Double discordance Conduction system

- Second AV node anterior
 - on the right side of the IA septum
 - on the anterior edge of mitral valve
- His bundle
 - enters the left ventricle between the MV and the PV
 - Courses in front of the PV annulus then on its left side before going down
 - In case of VSD, the His bundle is anteriorly located



Double discordance Conduction system



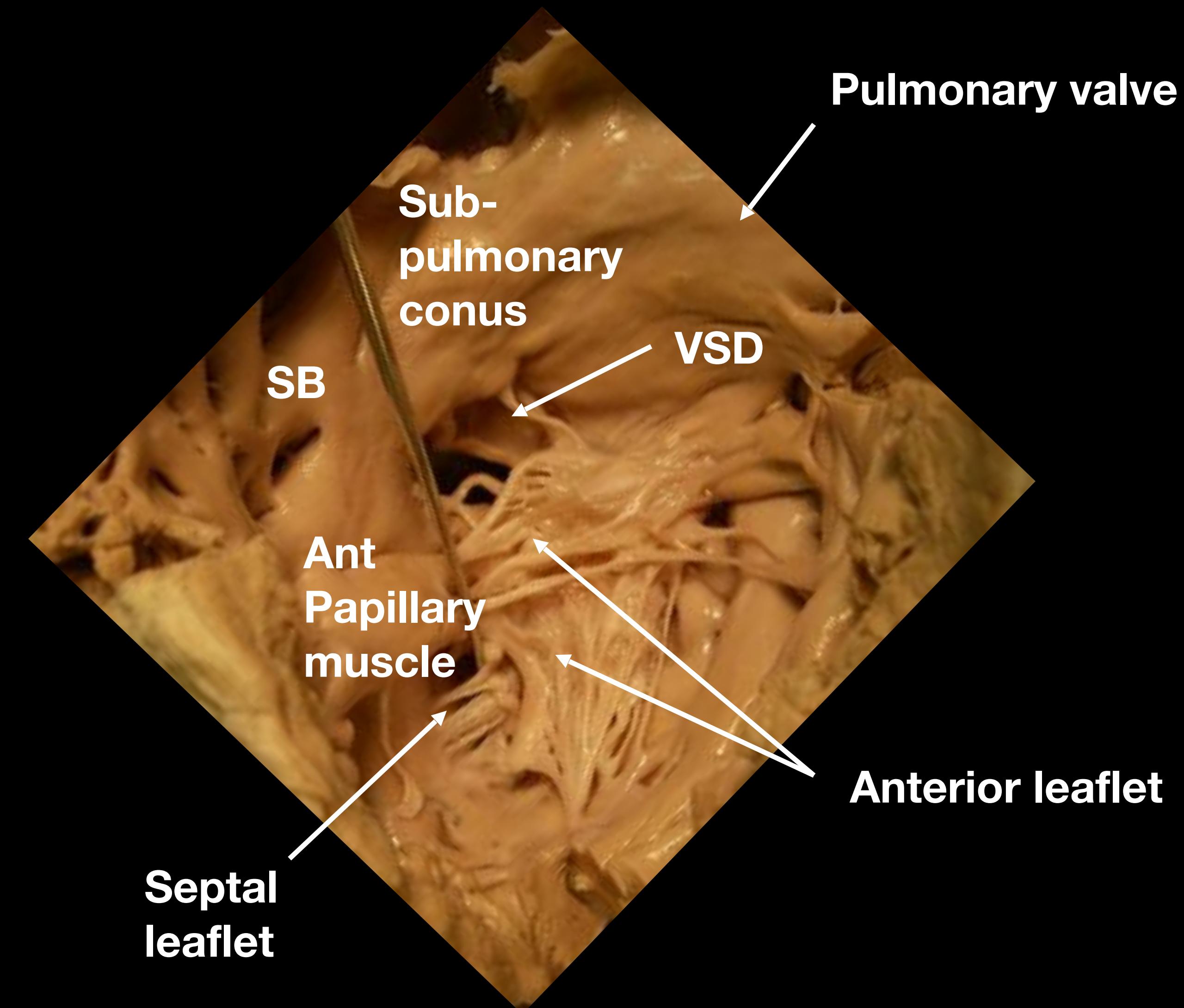
Double discordance Conduction system

- **Simple rules**
 - situs solitus : anterior node
 - situs inversus : posterior node
- **Many exceptions**
 - . alignment atrial septum/ventricular septum
 - . situs / position of cardiac apex
 - . 1 or 2 nodes – potential sling

Double discordance

The tricuspid valve

- Abnormal in more than 90% of cases
- Ebstein or dysplastic
- M3C collection
 - Ebstein 6/21 (29%)
 - dysplastic 2/21
 - straddling 7/21
 - overriding 2/21
 - hypoplasia 1/21
 - normale 5/21



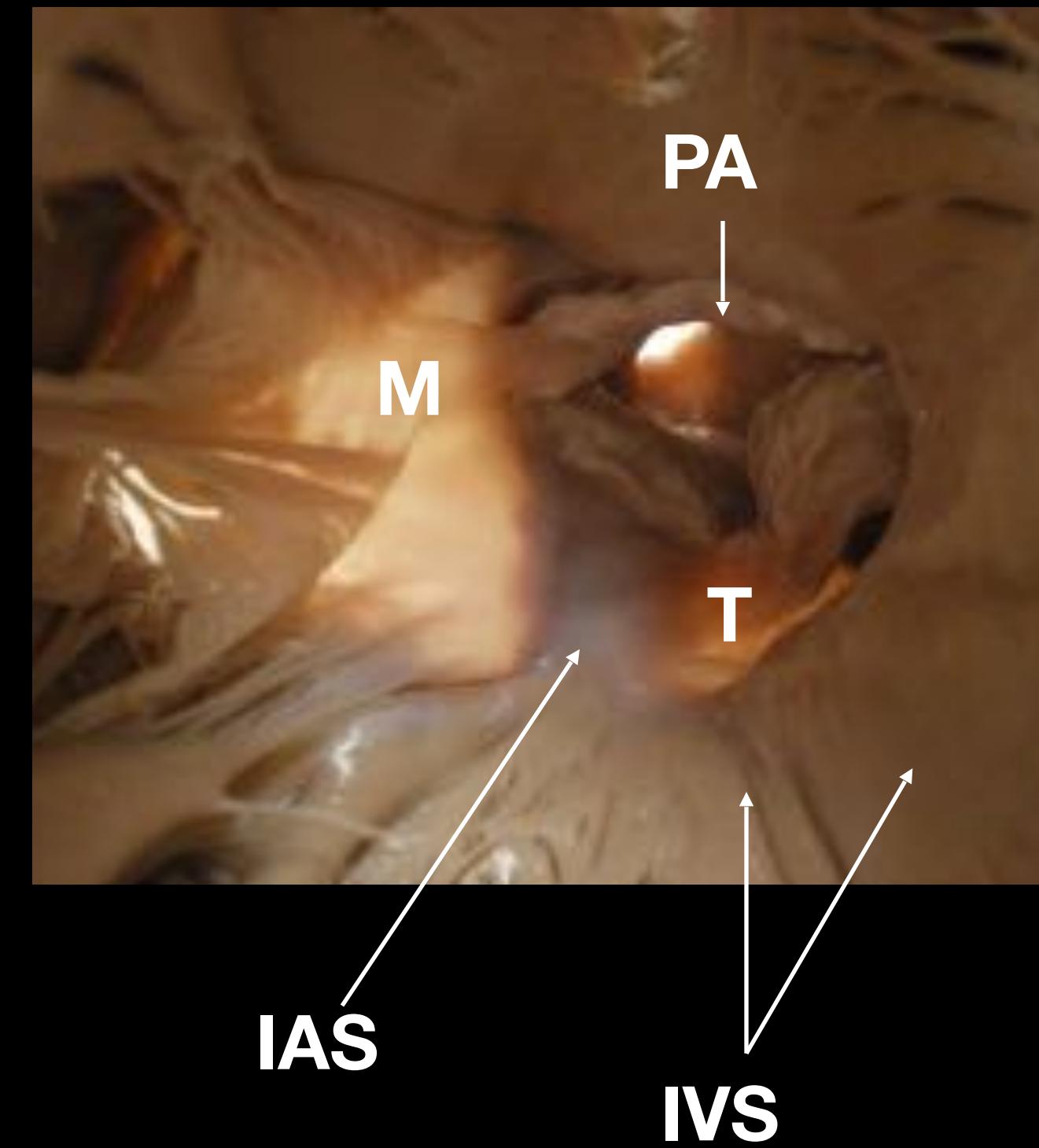
Double discordance Ebstein anomaly

- 30% in clinical series, 90% in anatomical series
- Similarities between Ebstein disease and « Ebstein » anomaly in DD
 - No delamination of septal and anterior leaflet
 - Maximal displacement of the tricuspid between the septal and posterior leaflet
 - Verticalisation of tricuspid valve orifice
- Différences
 - Atrial part of the right ventricle is thinner but always muscular
 - No dilatation of the tricuspid annulus
 - Cleft of the anterior leaflet can be seen

Double discordance

Anatomy summary

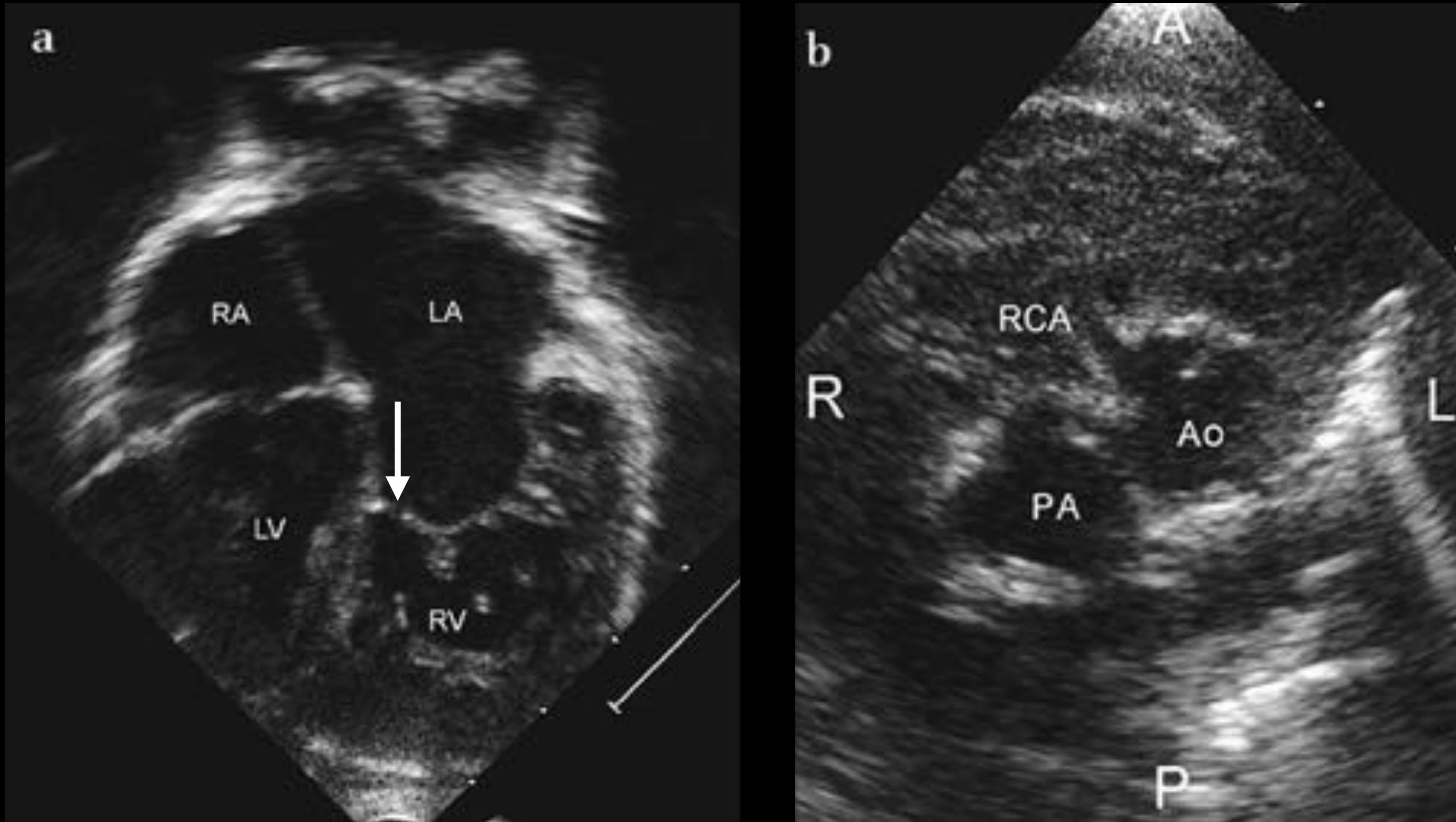
- **L laterality anomaly**
- **P primarily ventricular (looping)**
- **A associated with RV and tricuspid valve anomalies in 90% of cases**
- **M malalignment of septa creates**
 - **a abnormal His bundle course**
 - **L location of the VSD in the outlet**
 - **T trend to have straddling of tricuspid valve**



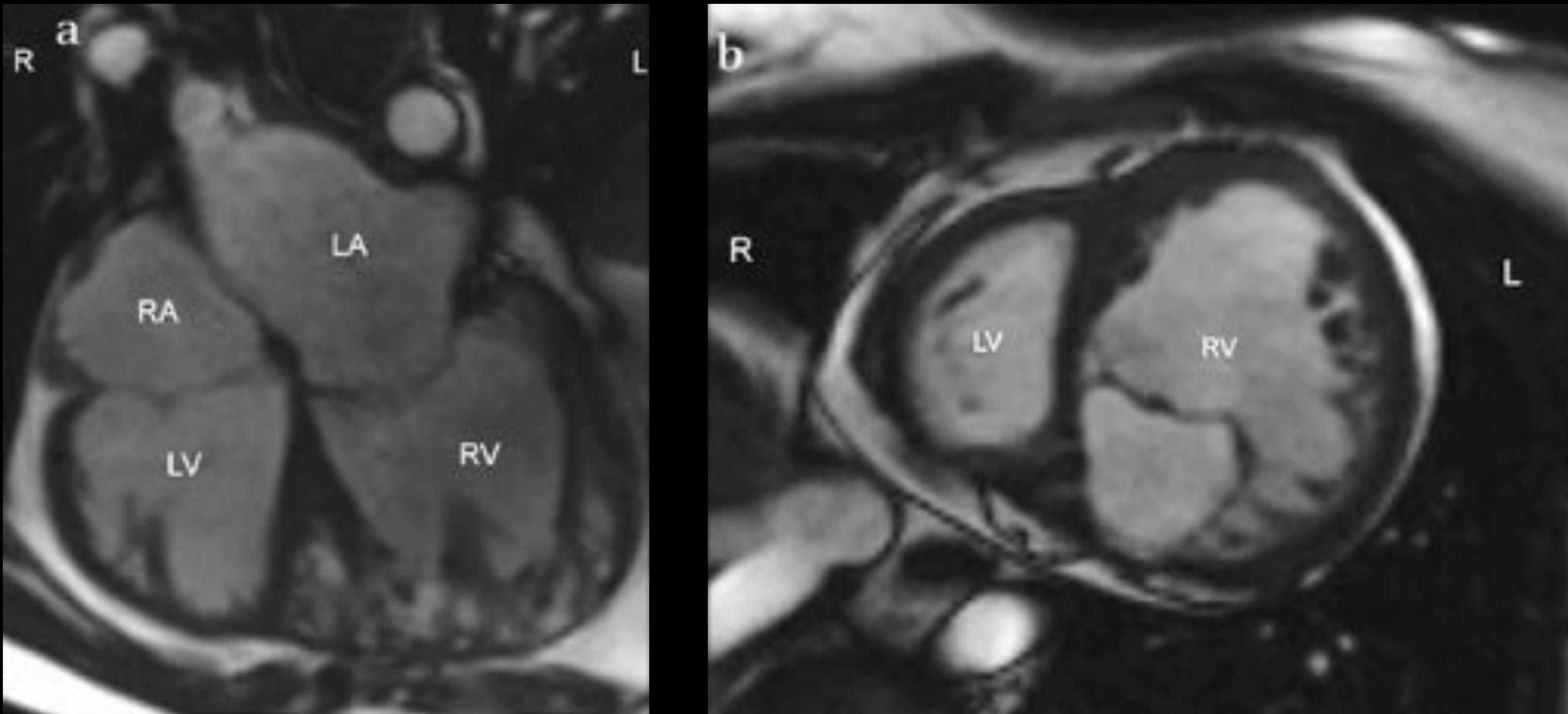
Double discordance

Imaging

Echocardiographic apical four chamber view demonstrating atrioventricular discordance.



Magnetic resonance imaging (MRI) four chamber view demonstrating atrioventricular discordance.



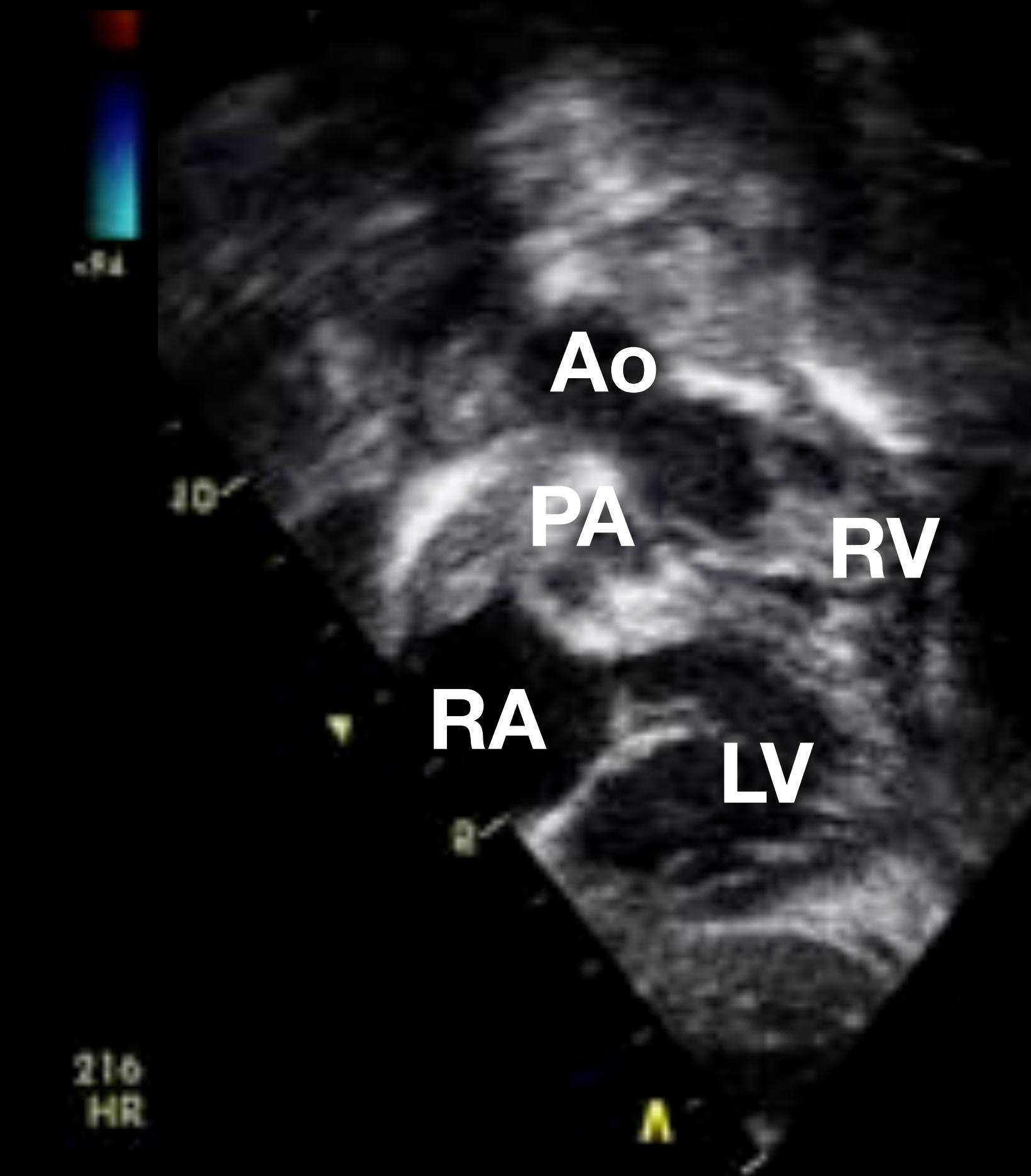
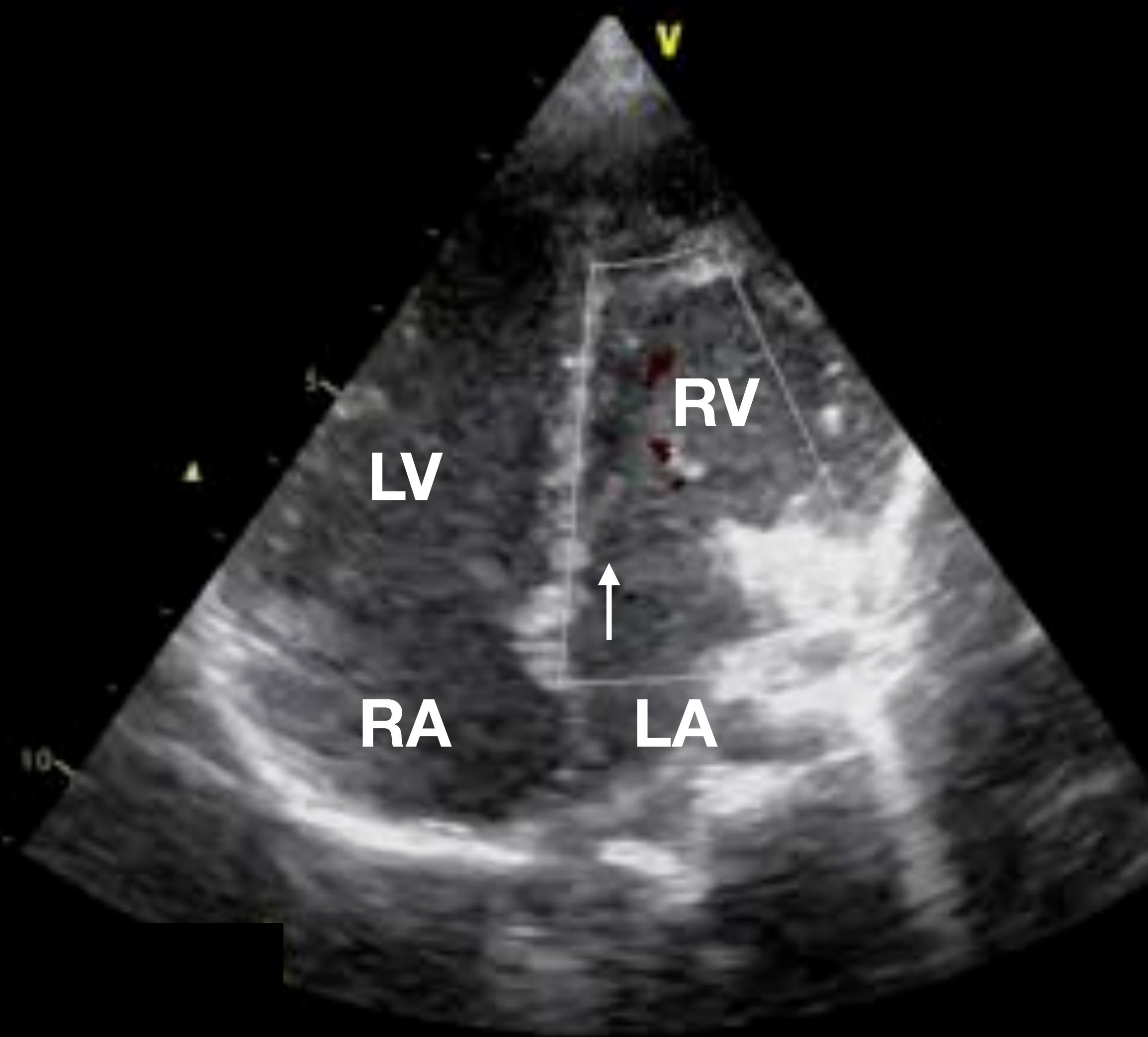
Hornung T S , and Calder L Heart 2010;96:1154-1161

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Heart

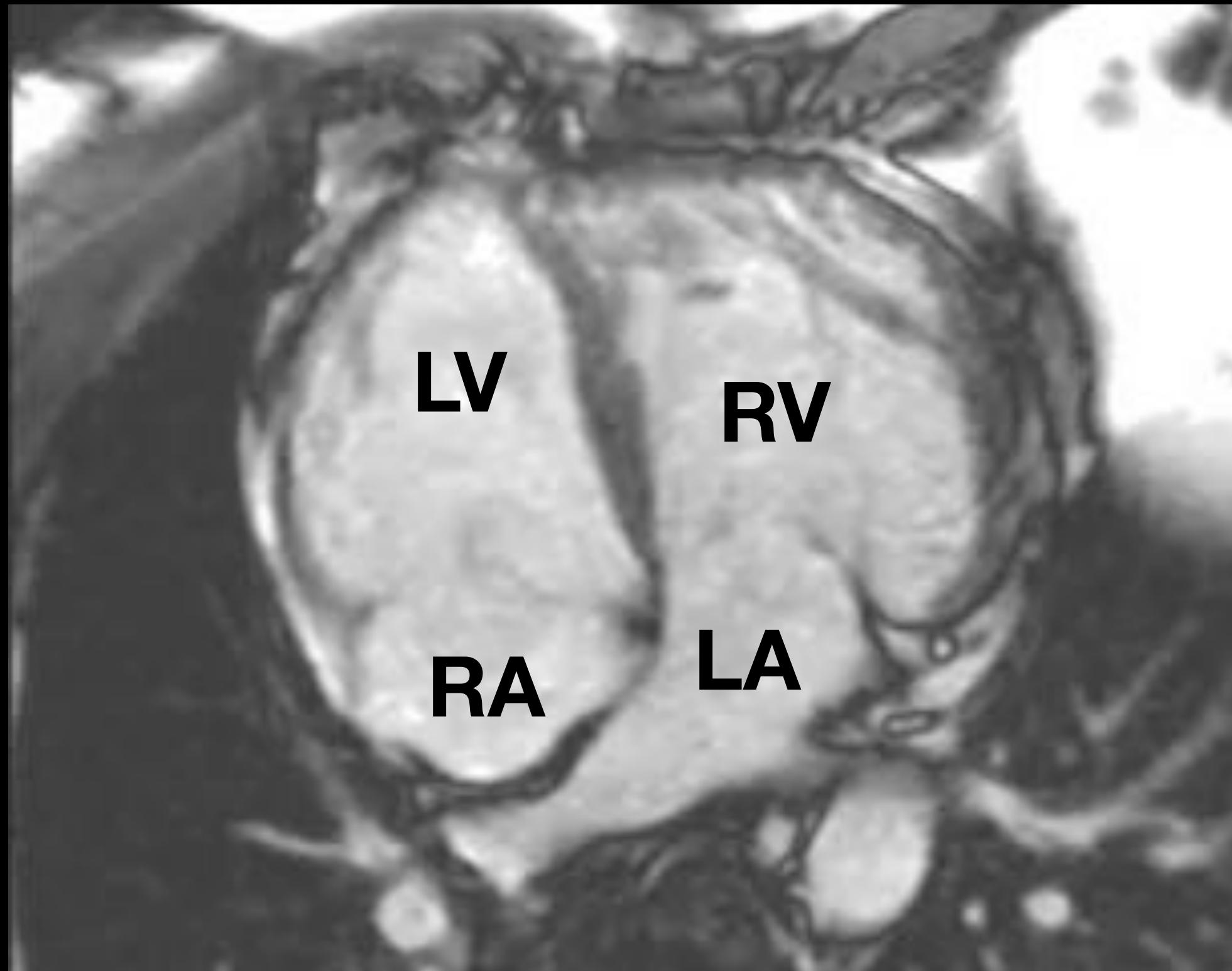
Congenitally corrected transposition of the great arteries

Double discordance

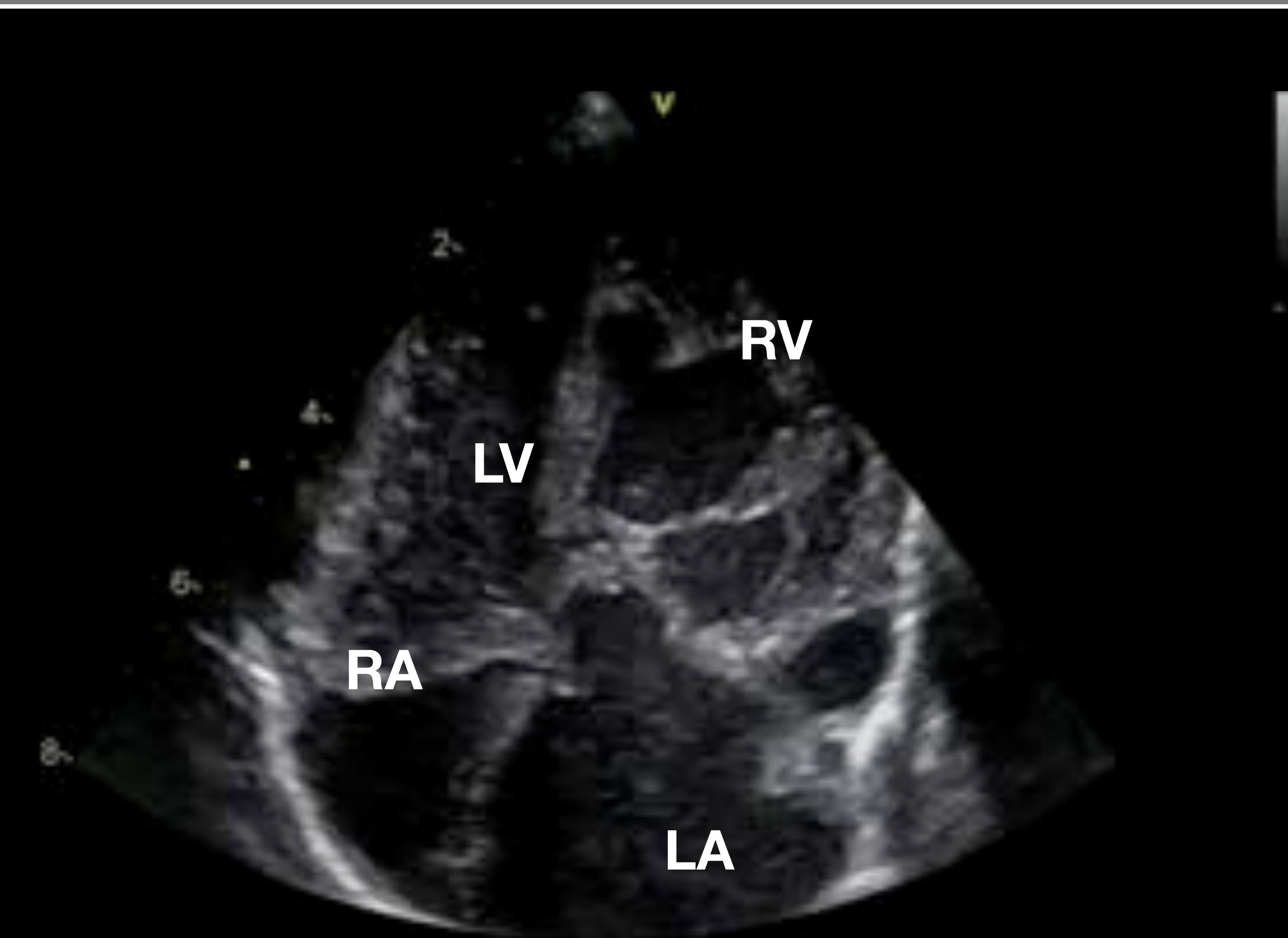


Congenitally corrected transposition of the great arteries

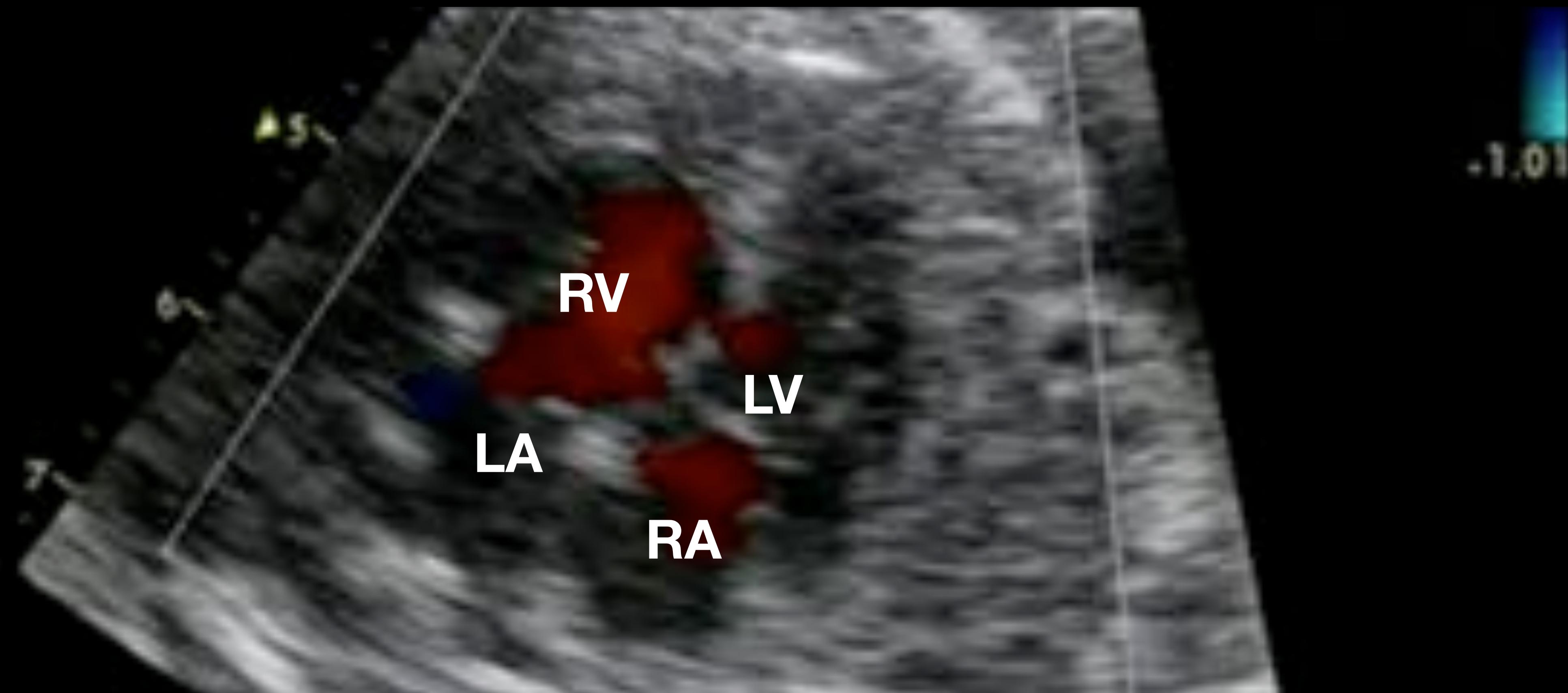
Double discordance



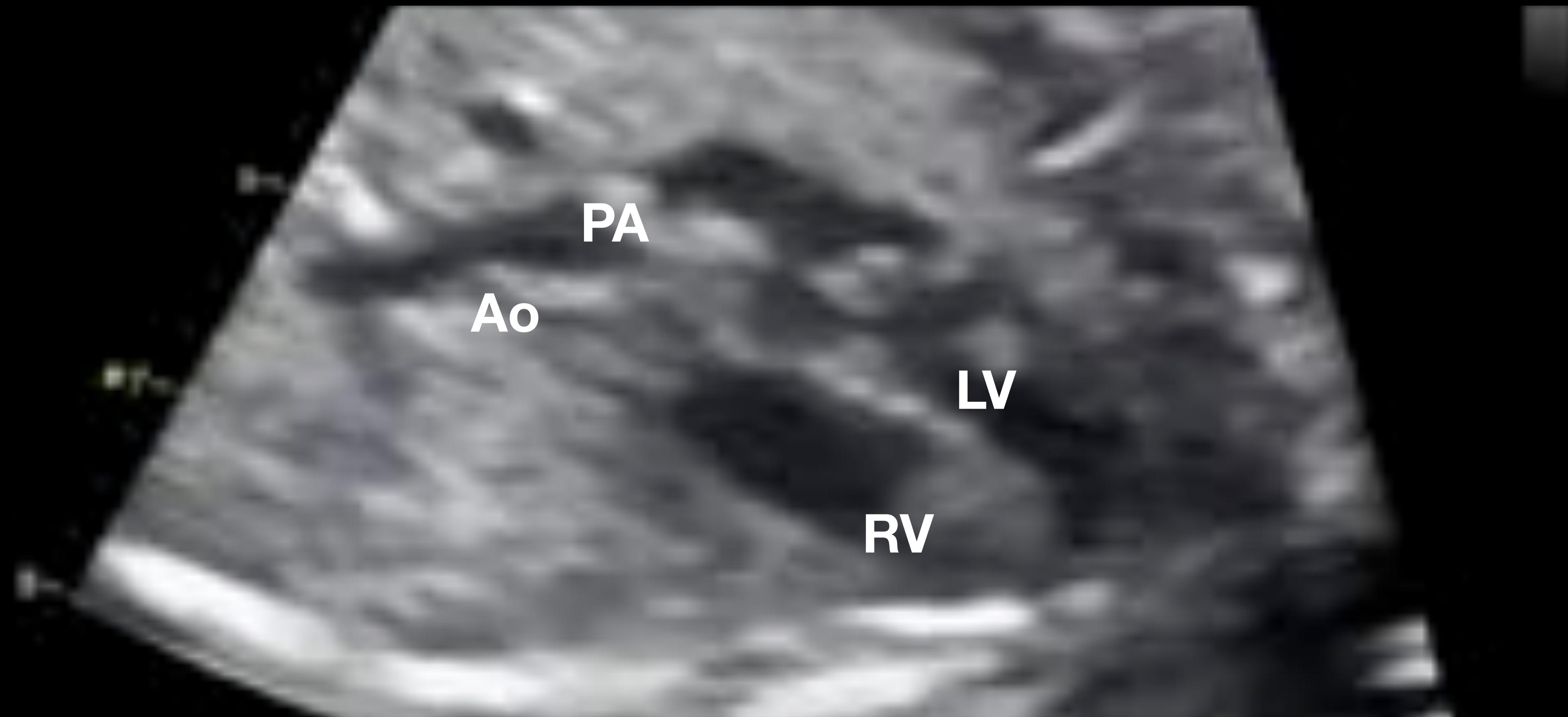
**Congenitally corrected transposition of the great arteries
Double discordance- Four chamber view**



**Congenitally corrected transposition of the great arteries
Double discordance- Foetal AV discordance**

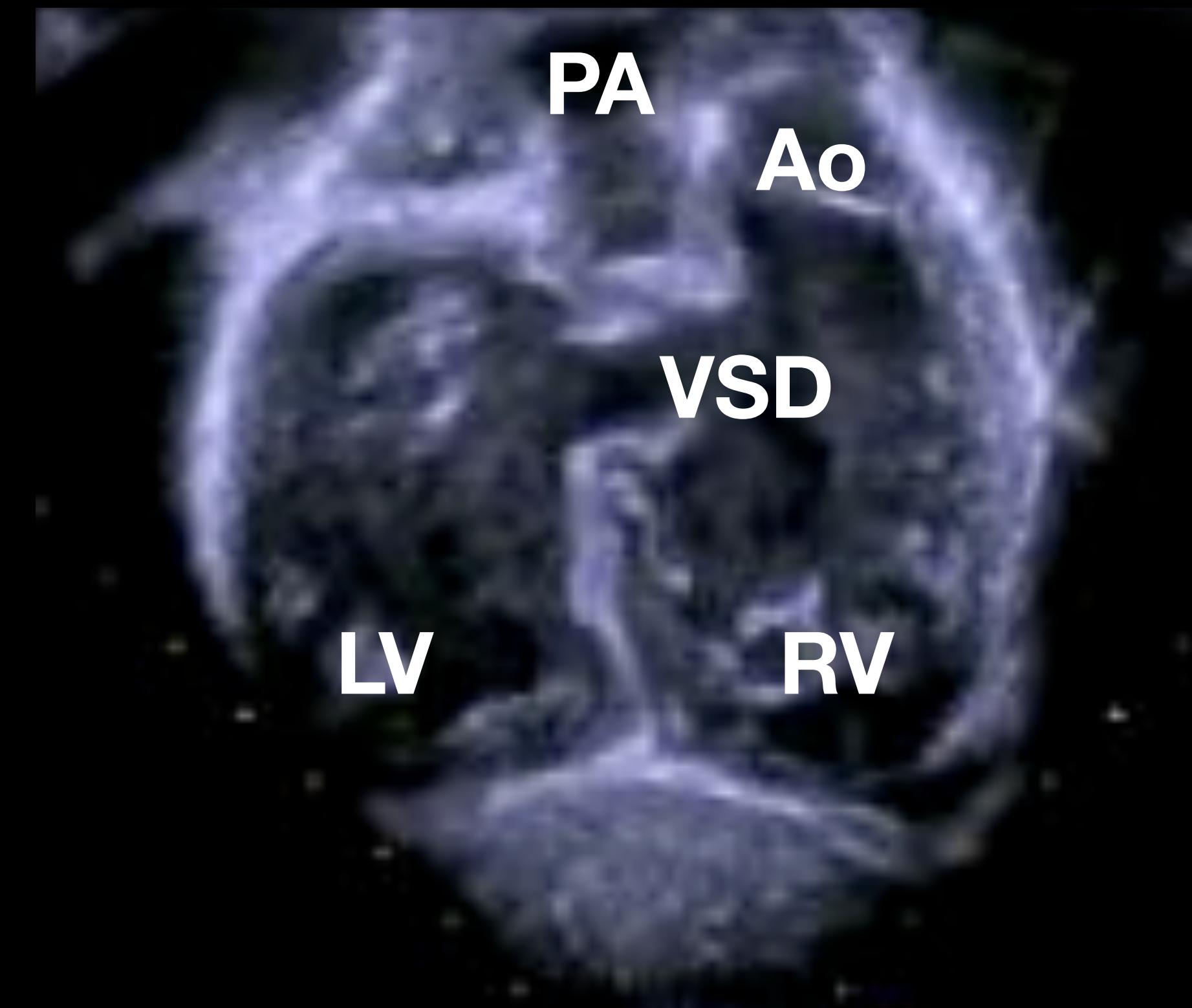


**Congenitally corrected transposition of the great arteries
Double discordance- Foetal vaisseaux**



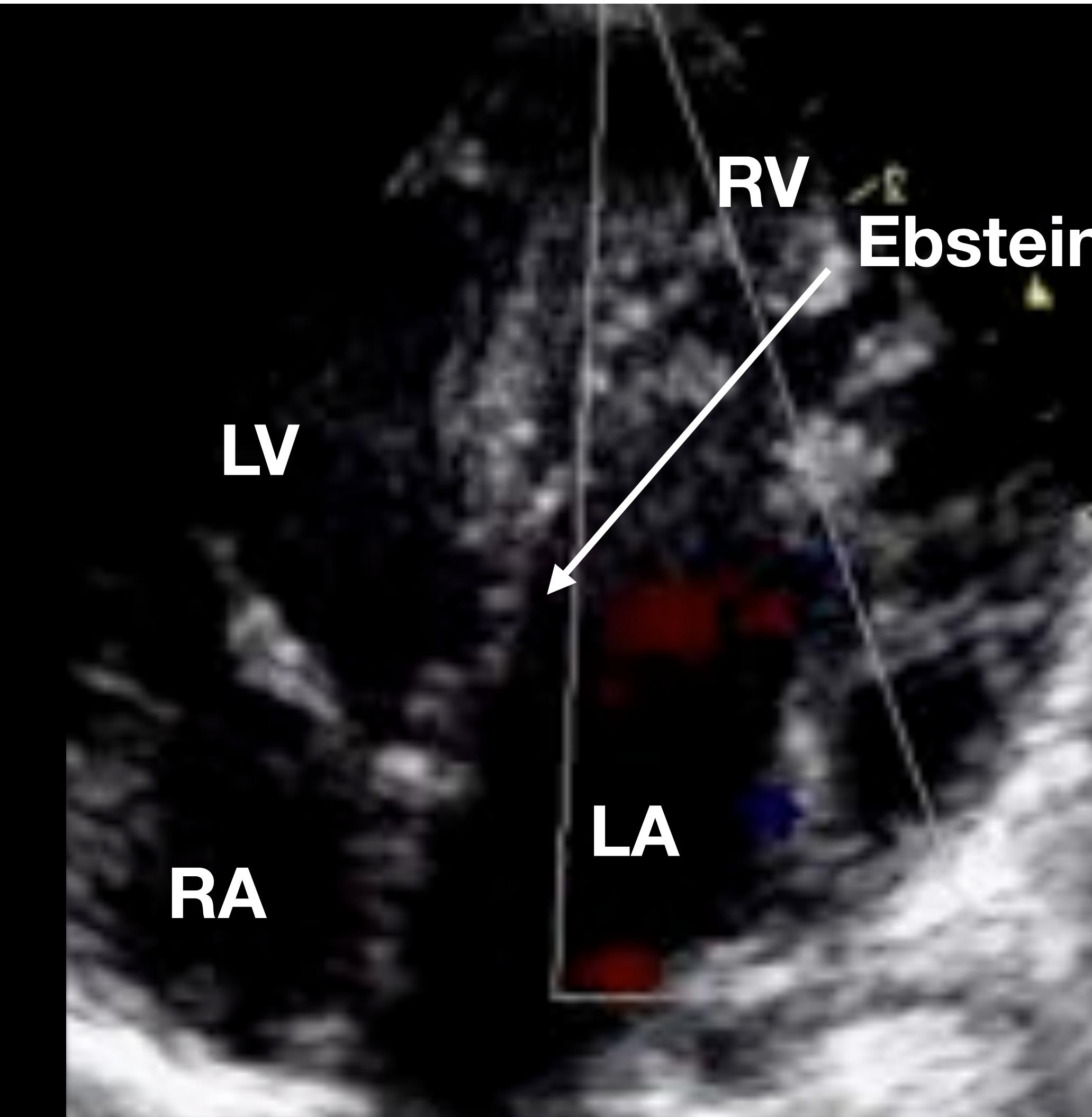
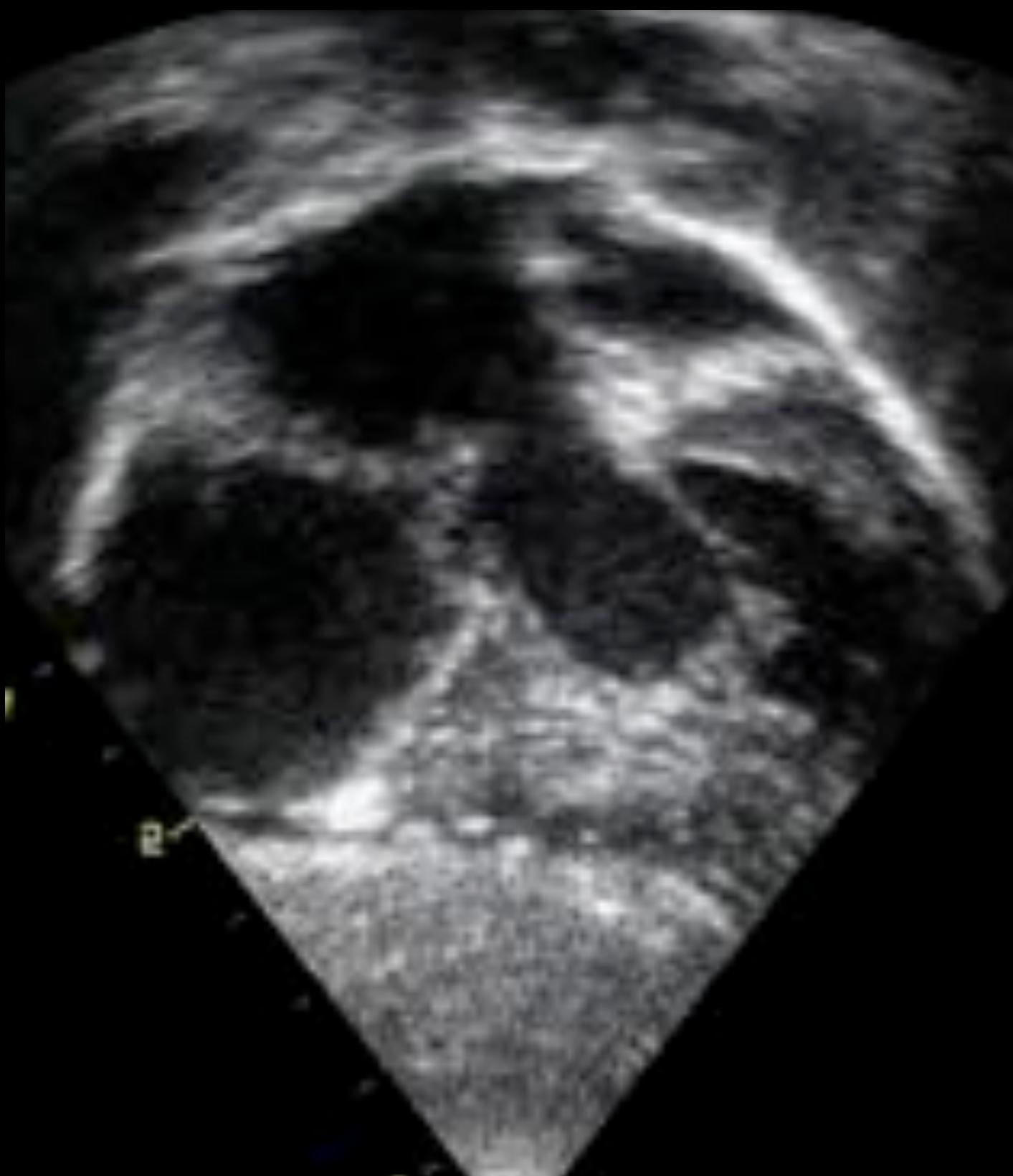
Congenitally corrected transposition of the great arteries

Double discordance

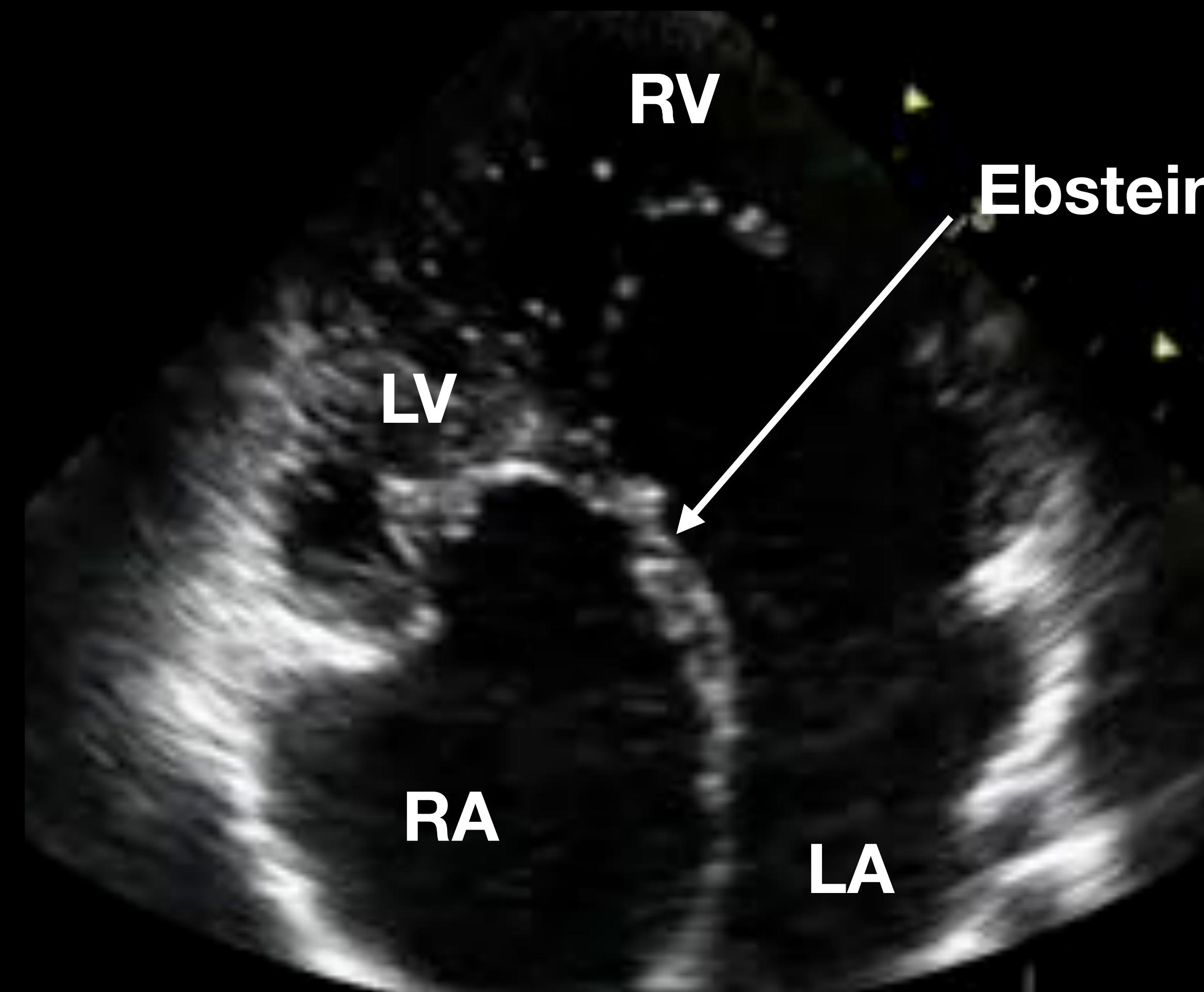


DD-VSD-PS

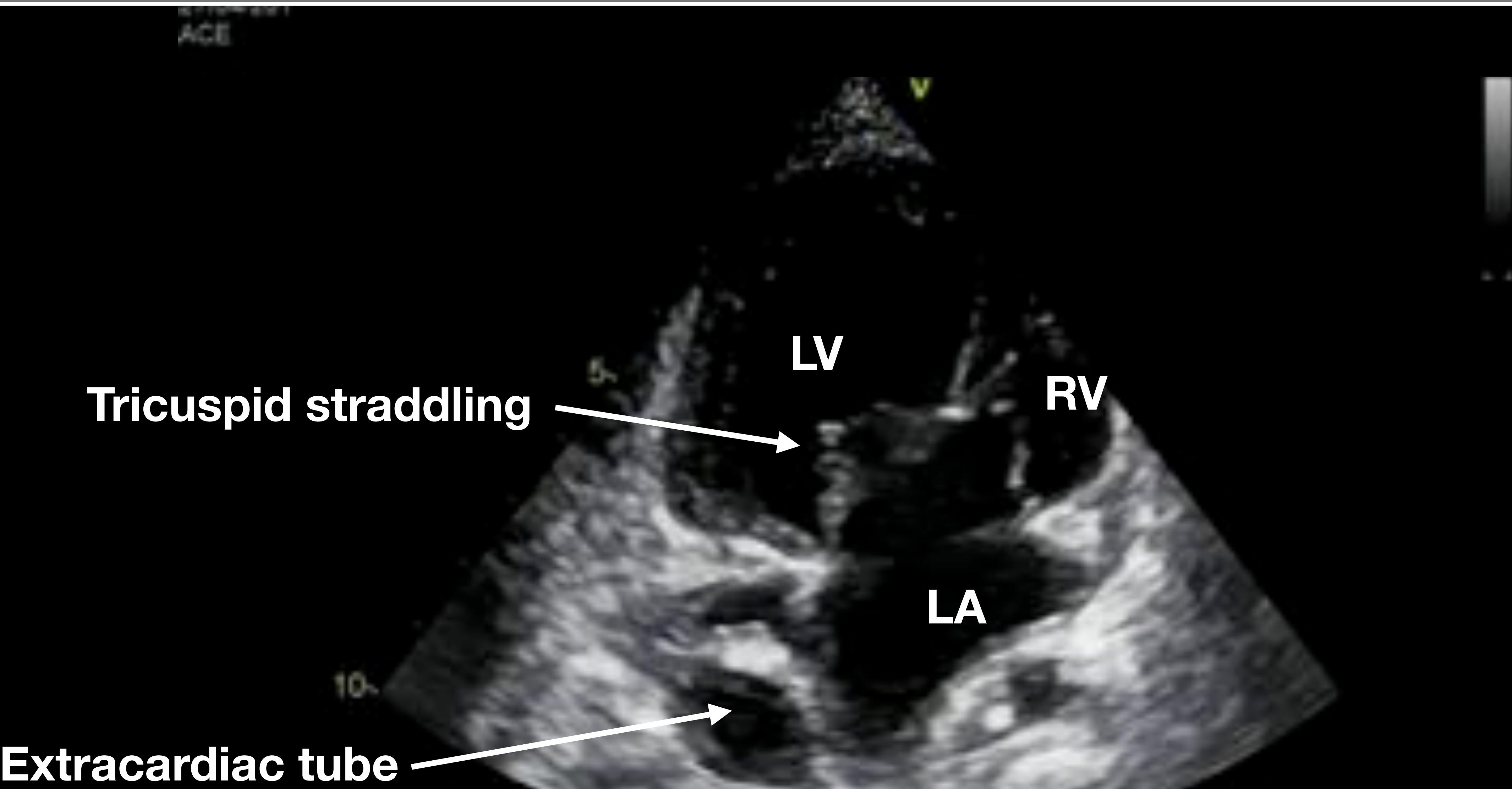
Congenitally corrected transposition of the great arteries
Double discordance - Ebstein



**Congenitally corrected transposition of the great arteries
Double discordance-Ebstein**



**Congenitally corrected transposition of the great arteries
Double discordance- Straddling tricuspid-Mitral atresia**



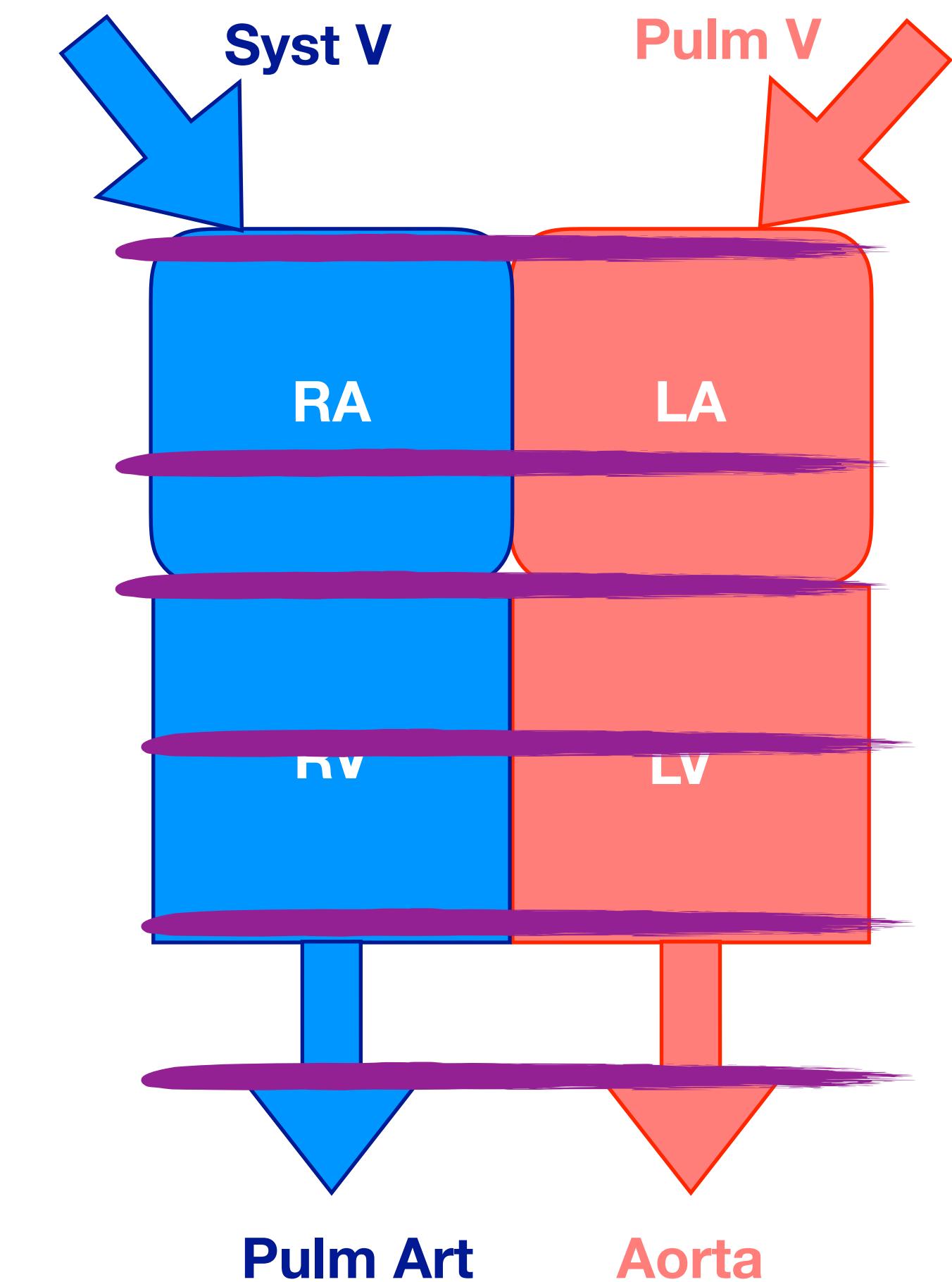
Double discordance
Clinical presentation
&
Natural history

The EPICARD study

EPIdémiologie des enfants ou fœtus ayant une CARDiopathie congénitale

Anatomic and Clinical Classification of Congenital Heart Diseases ACC-CHD

ACC-CHD categories	Examples
Heterotaxy	Heterotaxy syndromes
Anomalies of venous connections	Total anomalous pulmonary venous return
Anomalies of atria	Atrial septal defect
Anomalies of AV junction and AV valves	Atrioventricular septal defect
Complex anomalies of AV junction	Double discordance
Functionally univentricular heart	Hypoplastic left heart syndrome
Ventricular septal defects	Perimembranous VSD
Anomalies of ventriculo-arterial connections	Transposition of the great arteries, DORV
Anomalies of extra pericardial trunks	Coarctation of the aorta
Congenital anomalies of coronary arteries	ALCAPA



Prevalence, pre- and post-natal diagnosis, and infant mortality of newborns with congenital heart defects:
A population-based study using the International Paediatric and Congenital Cardiac Code (IPCCC)
The EPICARD Study Group

Distribution of categories of CHD and associated anomalies

Total number of birth

= 317 538

Live births

= 314 022

**Total
2867 cases**

**2349
Live birth
82.0%**

**465 TOP
16.2%**

**53 IUFD
1.8%**

N = 2867

**1753 (61.1%)
Isolated CHD**

**393 (13.7%)
Chromosomal
anomalies**

**409 (14.3%)
Extracardiac
anomalies**

ACC-CHD categories	Total		Live births	
	% of chromosomal anomalies	% of extra cardiac anomalies	% of chromosomal anomalies	% of extra cardiac anomalies
Heterotaxy	0	24.3	0	25.0
Anomalies of venous connections	19.4	16.1	7.7	15.4
Anomalies of atria	9.9	19.8	7.5	19.0
Anomalies of AV junction and AV valves	57.3	12.7	43.1	13.8
Complex anomalies of AV junction	0	7.7	0	0
Functionally univentricular heart	15.8	19.6	8.3	20.8
Ventricular septal defects	9.3	11.1	3.9	11.0
Anomalies of ventriculo-arterial connections	10.7	18.8	4.5	14.1
Anomalies of extra pericardial trunks	15.9	31.2	3.2	26.4
Congenital anomalies of coronary arteries	0	0	0	0

Prevalence, pre- and post-natal diagnosis, and infant mortality of newborns with congenital heart defects
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The EPICARD Study Group

Proportion of prenatal diagnosis

All CHDs

ACC-CHD categories	% of prenatal diagnosis
All cases excluding chromosomal anomalies	25.6
All cases excluding chromosomal and other extra cardiac anomalies	23
All cases excluding chromosomal, other anomalies and simple VSD	40.2

In categories of CHDs

ACC-CHD categories	% of prenatal diagnosis (n)
Heterotaxy	89.2 (37)
Anomalies of venous connections	16.0 (25)
Anomalies of atria	4.3 (164)
Anomalies of AV junction and AV valves	67.0 (91)
Complex anomalies of AV junction	100.0 (13)
Functionally univentricular heart	92.5 (133)
Ventricular septal defects	9.6 (1353)
Anomalies of ventriculo-arterial connections	39.2 (503)
Anomalies of extra pericardial trunks	44.7 (143)
Congenital anomalies of coronary arteries	0 (9)

Specific CHDs

Type of CHD	% of prenatal diagnosis
Congenitally corrected transposition of the great	100
Functionally univentricular heart	92.5
TGA	84
DORV	98

Prevalence, pre- and post-natal diagnosis, and infant mortality of newborns with congenital heart defects:
A population-based study using the International Paediatric and Congenital Cardiac Code (IPCCC)
The EPICARD Study Group

Proportion of Termination of pregnancy

All CHDs : total 16%

ACC-CHD categories	% TOP
All cases excluding chromosomal anomalies	9.8
All cases excluding chromosomal and other extra cardiac anomalies	6.4
All cases excluding chromosomal, other anomalies and simple VSD	14.0

In categories of CHDs

ACC-CHD categories	% TOP
Heterotaxy	75.7
Anomalies of venous connections	16.1
Anomalies of atria	4.4
Anomalies of AV junction and AV valves	42.7
Complex anomalies of AV junction	46.2
Functionally univentricular heart	62.7
Ventricular septal defects	5.7
Anomalies of ventriculo-arterial connections	18.5
Anomalies of extra pericardial trunks	23.5
Congenital anomalies of coronary arteries	0

Epidemiology

- 1/33.000 life births
- 0.05% of CHD

Circumstances of diagnosis

- Fortuitously in isolated DD
- Chest-X ray (dextro or mesocardia)
- ECG
- Echocardiography for murmur
- AV Block
 - 1/3 of adult patients
 - May be present in fetus: incidence +2% per year
- Autopsy

Clinical examination

- Loud second heart sound : anterior aorta
- VSD, Pulmonary stenosis or tricuspid regurgitation murmur
- Cyanosis when VSD+ severe Pulm. stenosis
- Heart failure

Double discordance

ECG

- absence of Q waves in the lateral precordium (V4-6) or in aVL
 - physiologic Q waves in these leads often referred to as "septal" Q waves
- prominent Q waves in right precordium (V1) / QR in V1
- third degree atrioventricular (AV) conduction block

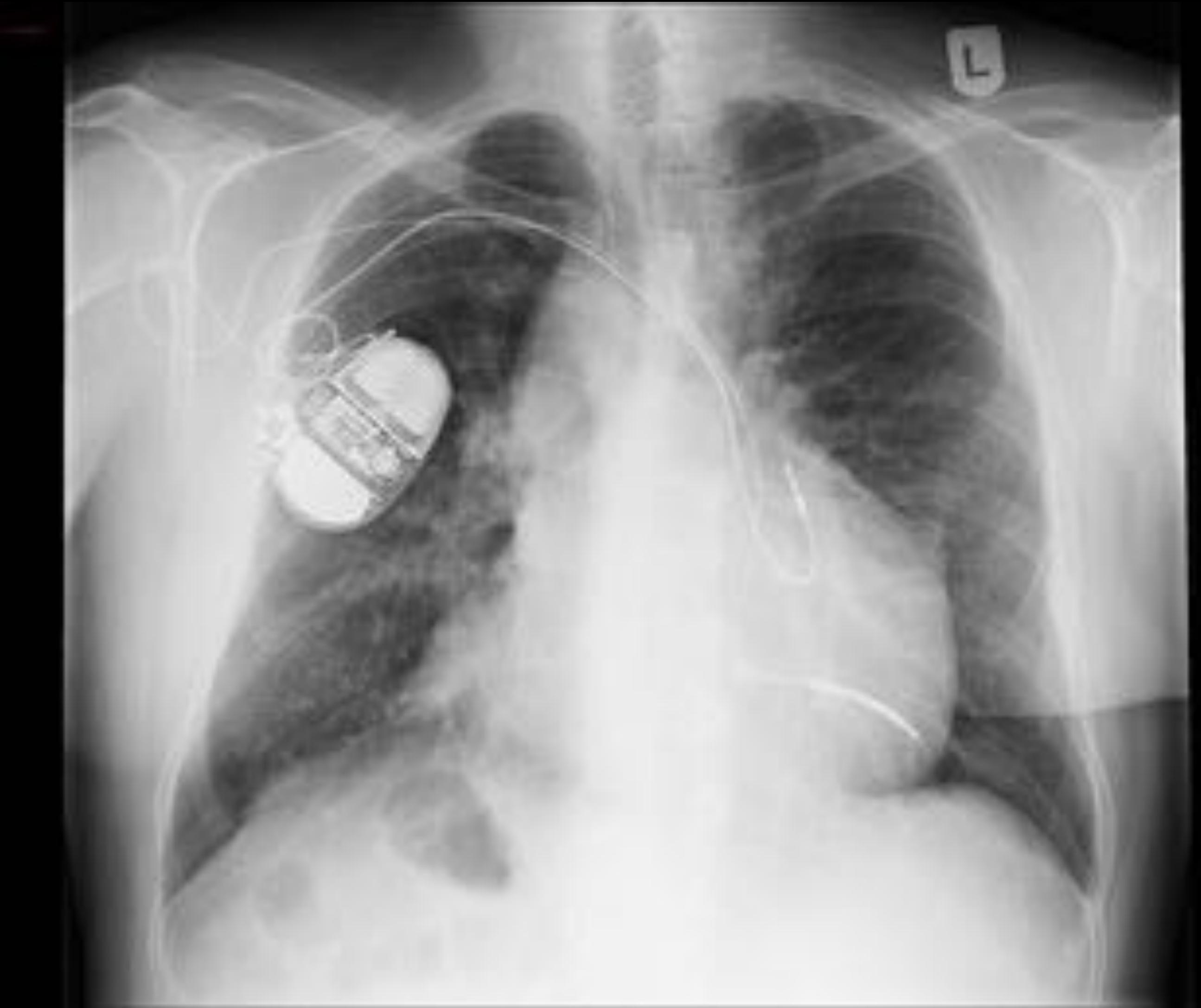


Double discordance ECG



Double discordance

Chest X-ray



Natural history

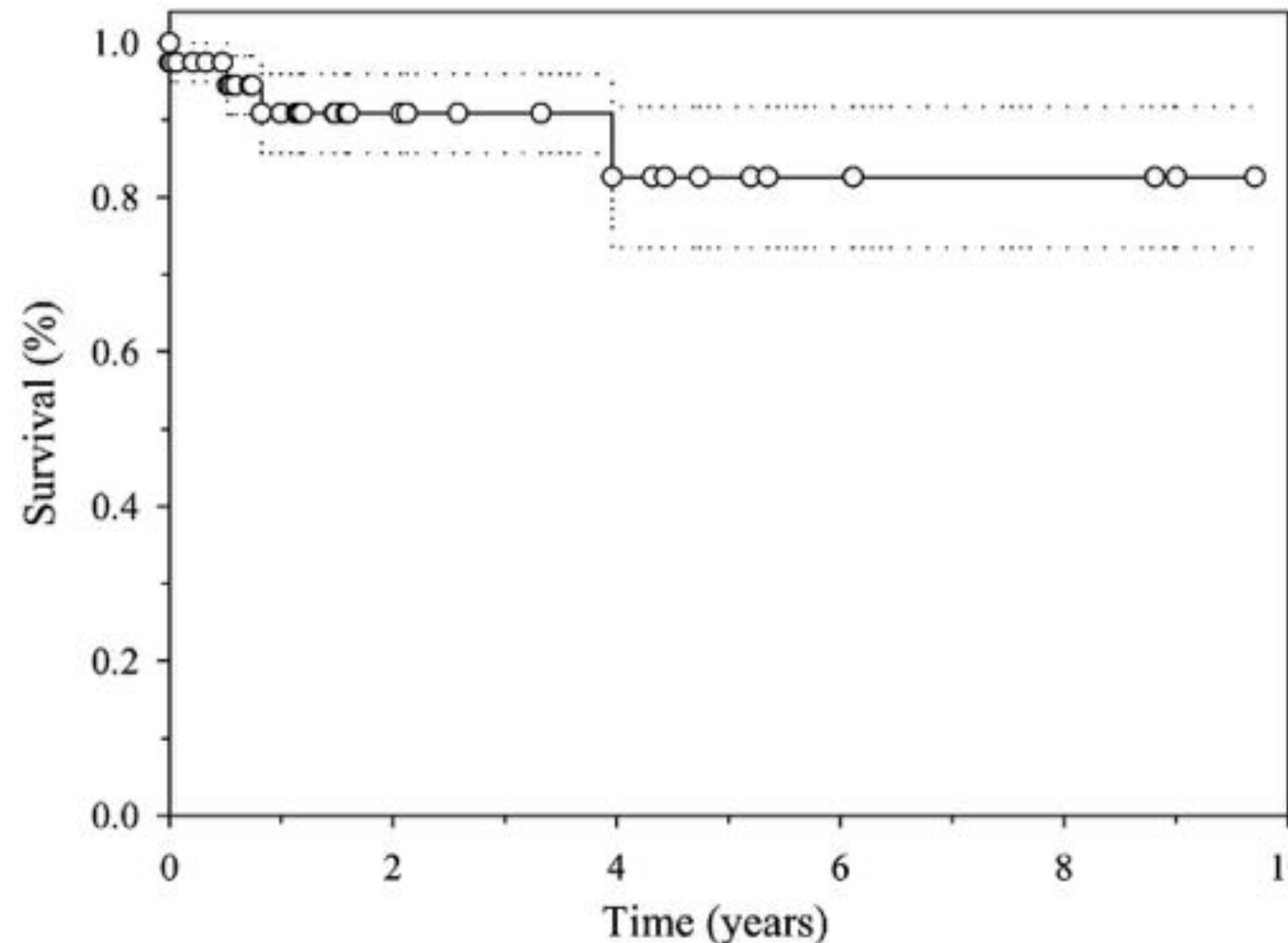
- AV block
 - 10-15% in adolescent
 - 30% in adults after 30 years
- Right ventricular dysfunction
- Tricuspid regurgitation

Long term survivors: exceptions

- 51 years Int J Cardiol 2007
- 70 years Hellenic J Cardiol 2005
- 72 and 80 years Arch Mal Cœur Vaiss 1986
- 76 years Ujeskr Laeger 2008
- 80 years Heart 1998

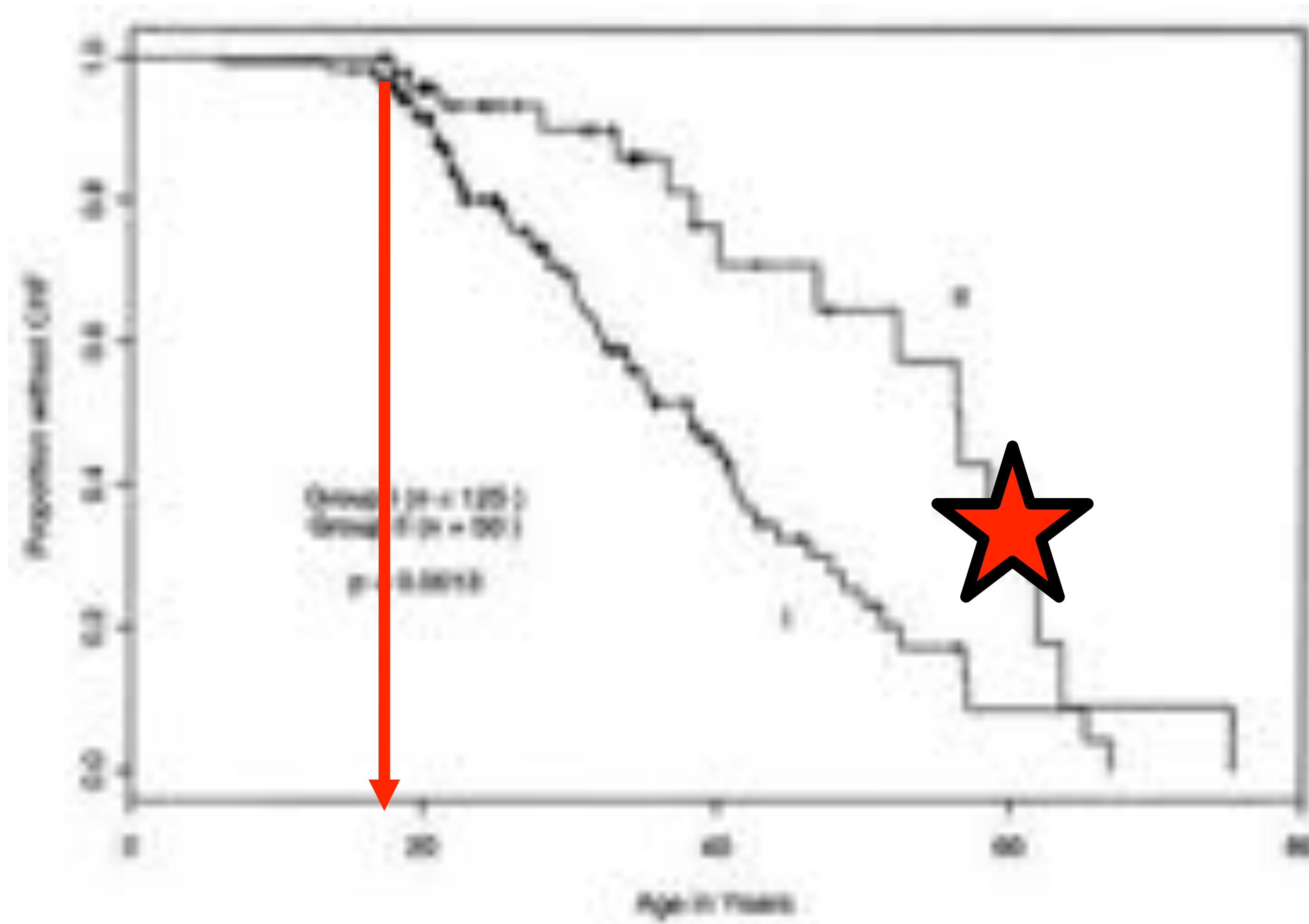
Double discordance - Survival in childhood

Kaplan-Meier survival curve of events include death and transplantation



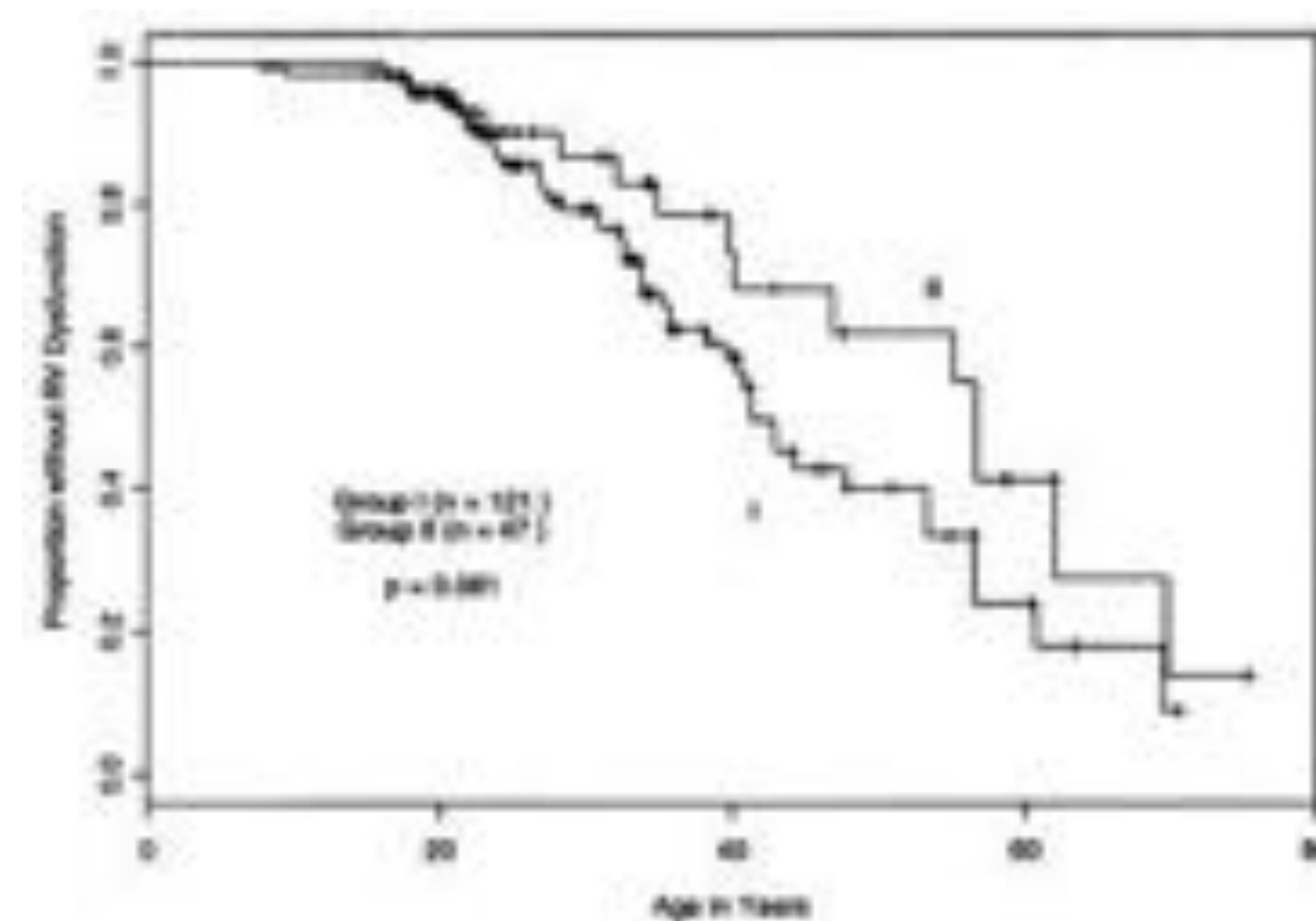
Double discordance - Survival in adulthood

Survival without heart failure



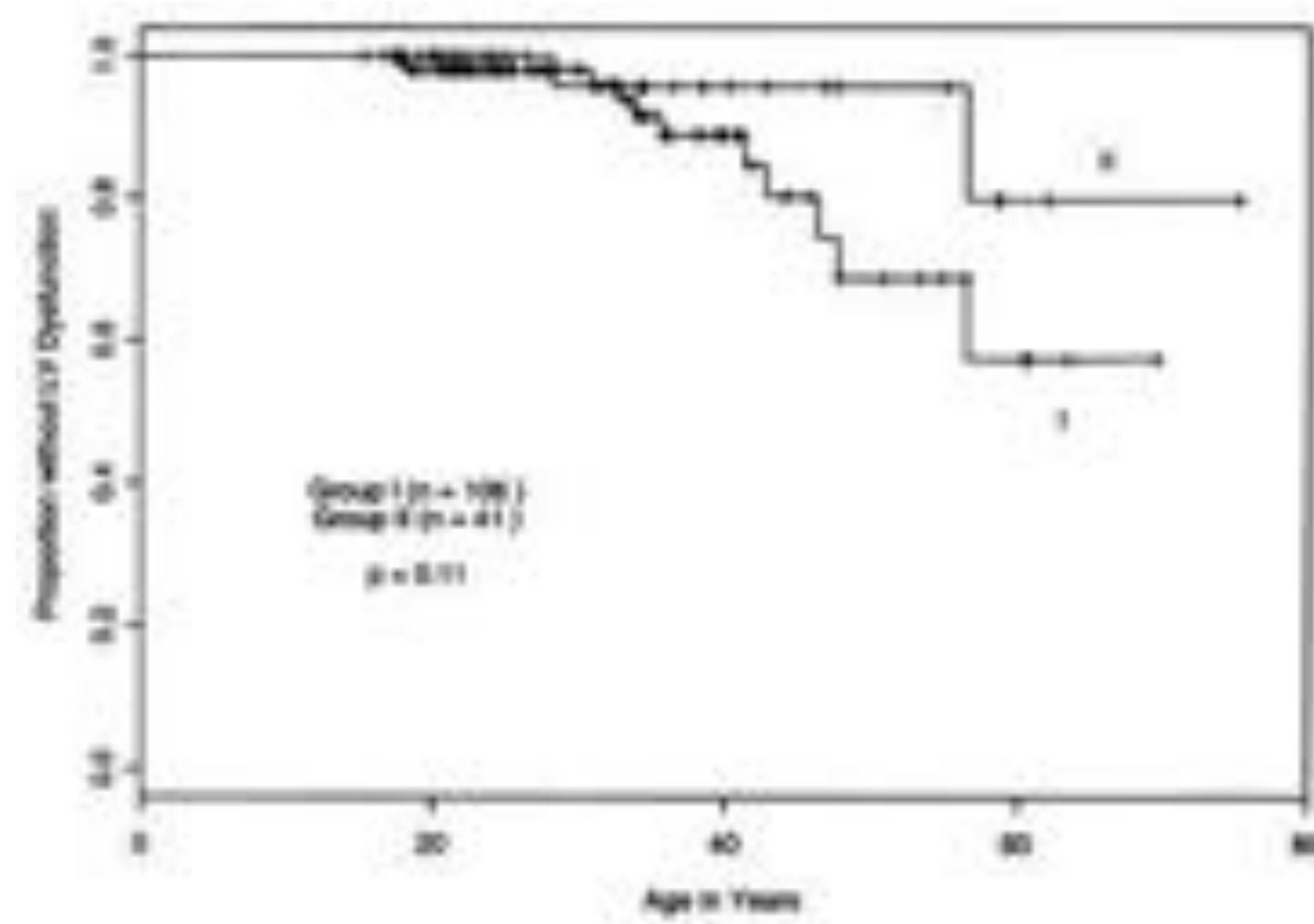
Double discordance

Freedom of Right (systemic) ventricular dysfunction

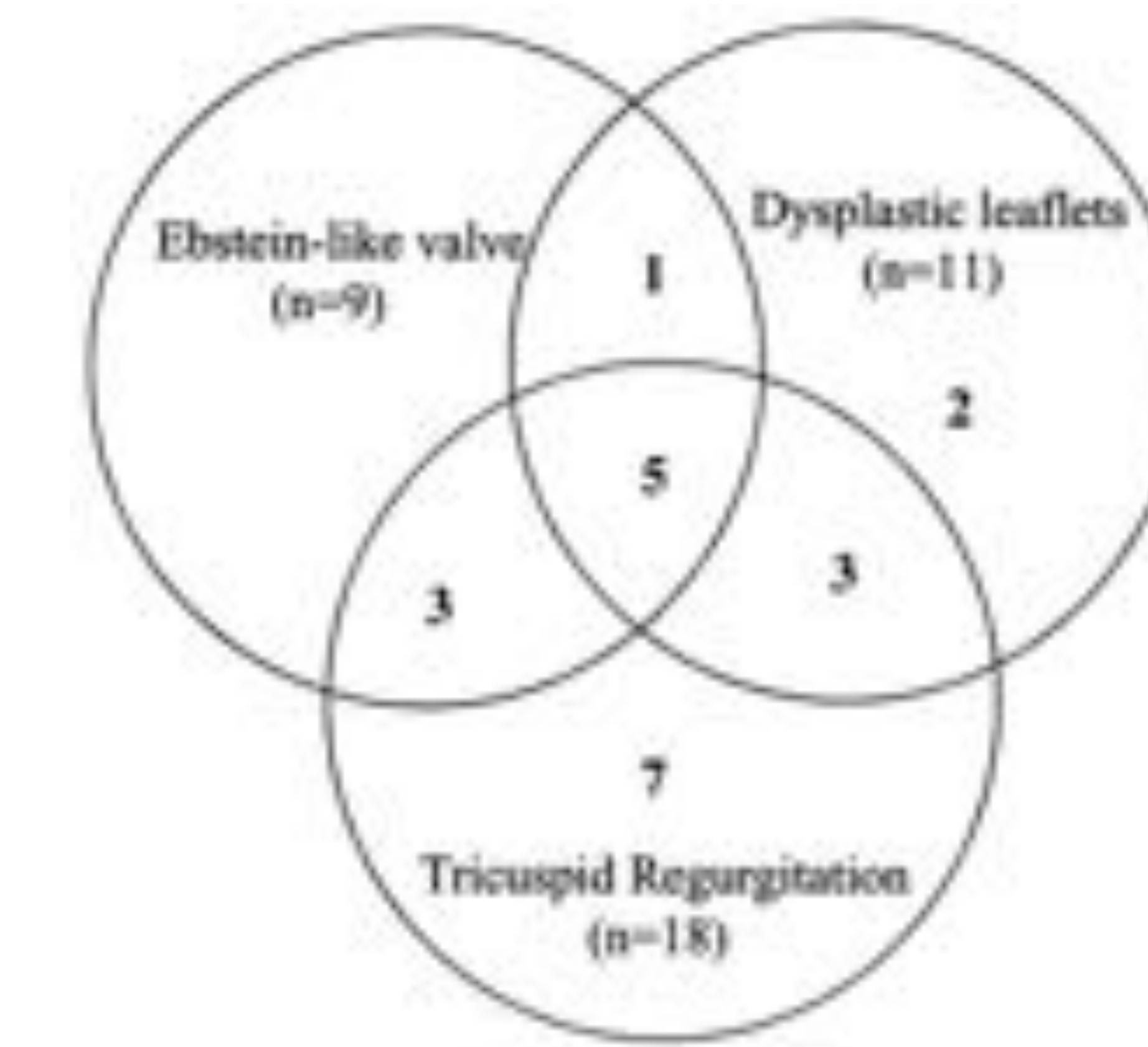


Double discordance

Freedom from Left ventricular dysfunction



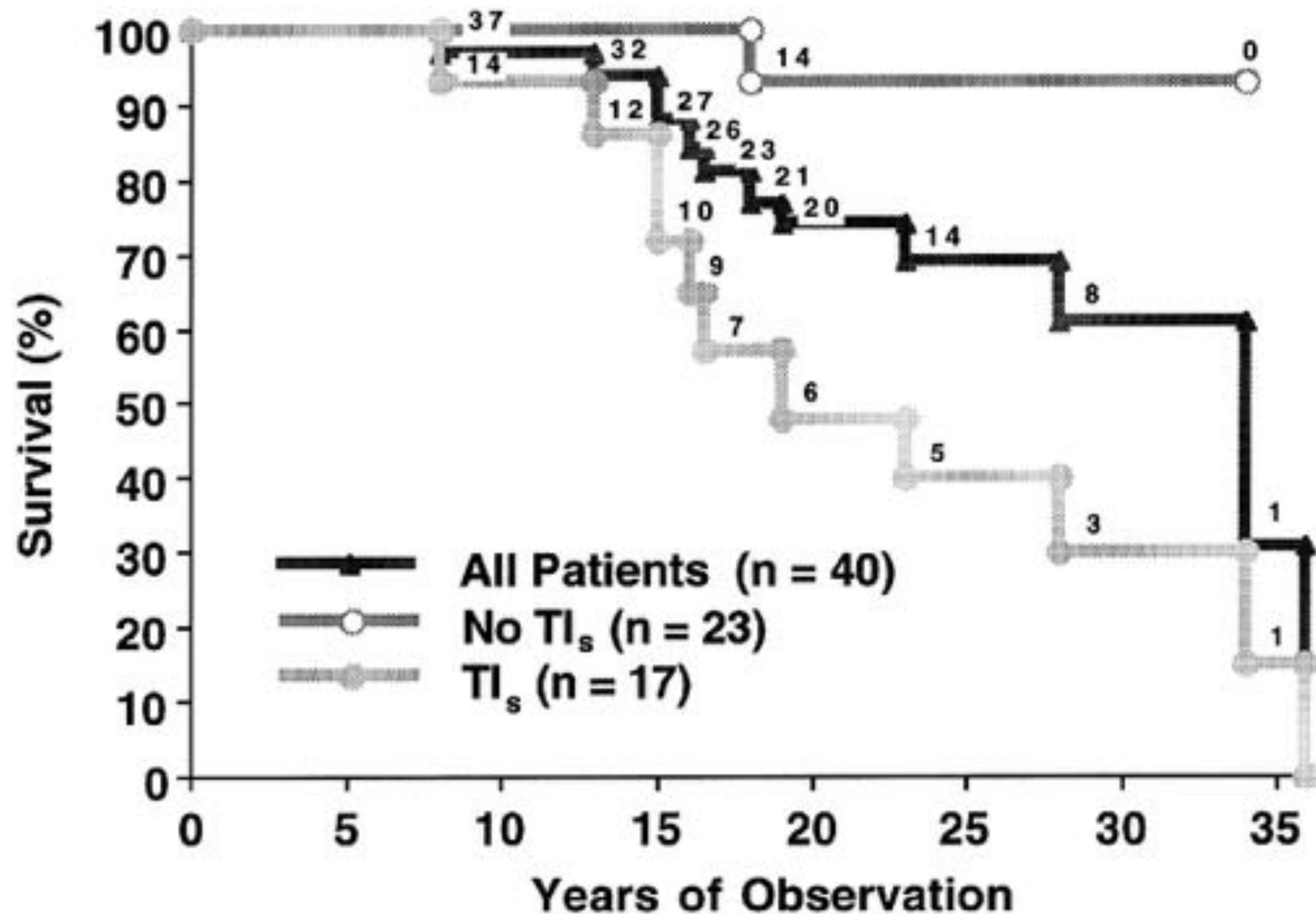
Double discordance Tricuspid valve



- 66% morphological anomalies in ccTGA (Acar 1998)
- 59% have TR > 3/4 in adulthood (Beauchesne 2002)
- TR is a major risk factor for death (Prieto 1998)
- TR and RV dysfunction are closely linked (Graham 2000)

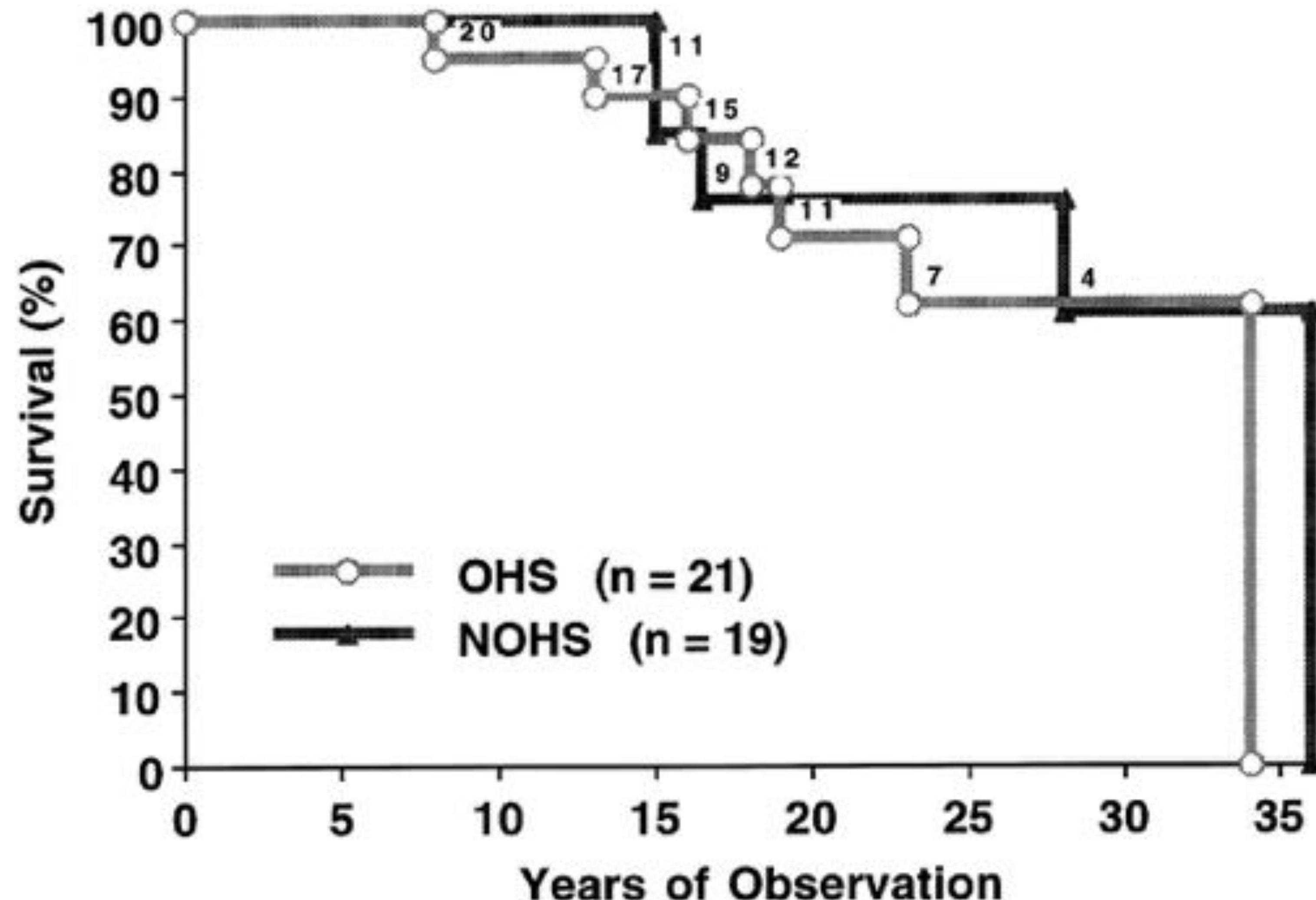
Double discordance - Survival and tricuspid regurgitation

Survival for all 40 patients with CCTGA: tricuspid regurgitation



Double discordance - Survival and tricuspid regurgitation

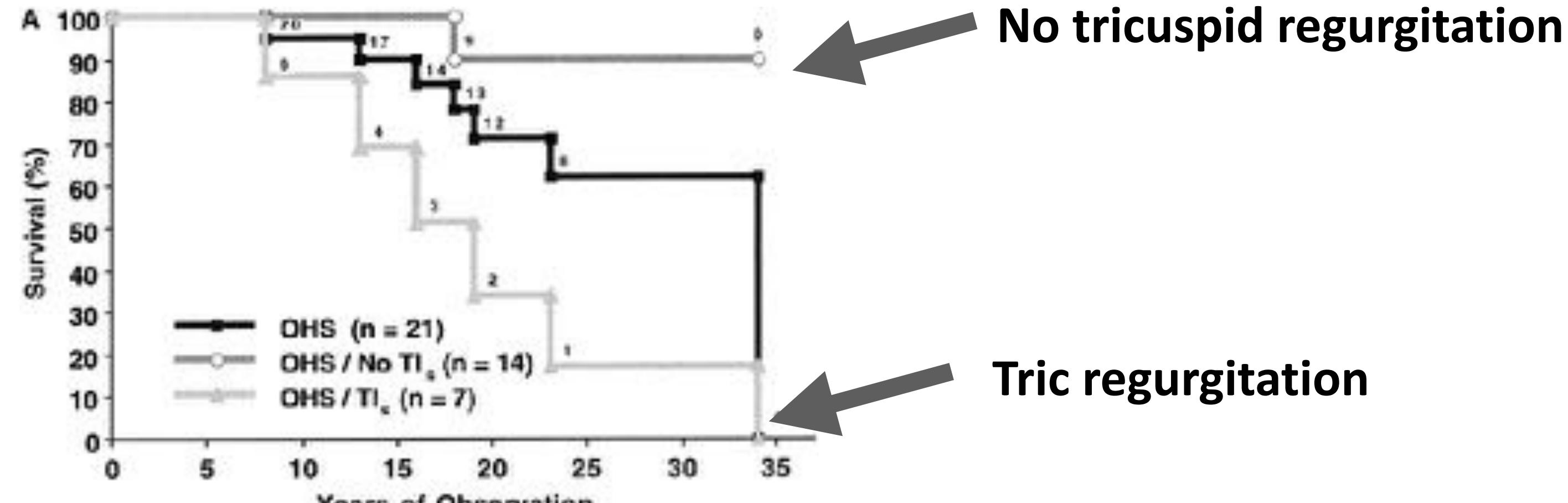
Survival for 21 patients with CCTGA who underwent open heart surgery for associated lesions and for 19 unoperated patients



Double discordance - Survival and tricuspid regurgitation

Survival for A, 21 patients who underwent open heart surgery, and B, 19 unoperated patients broken down into those without TIs and those with TIs

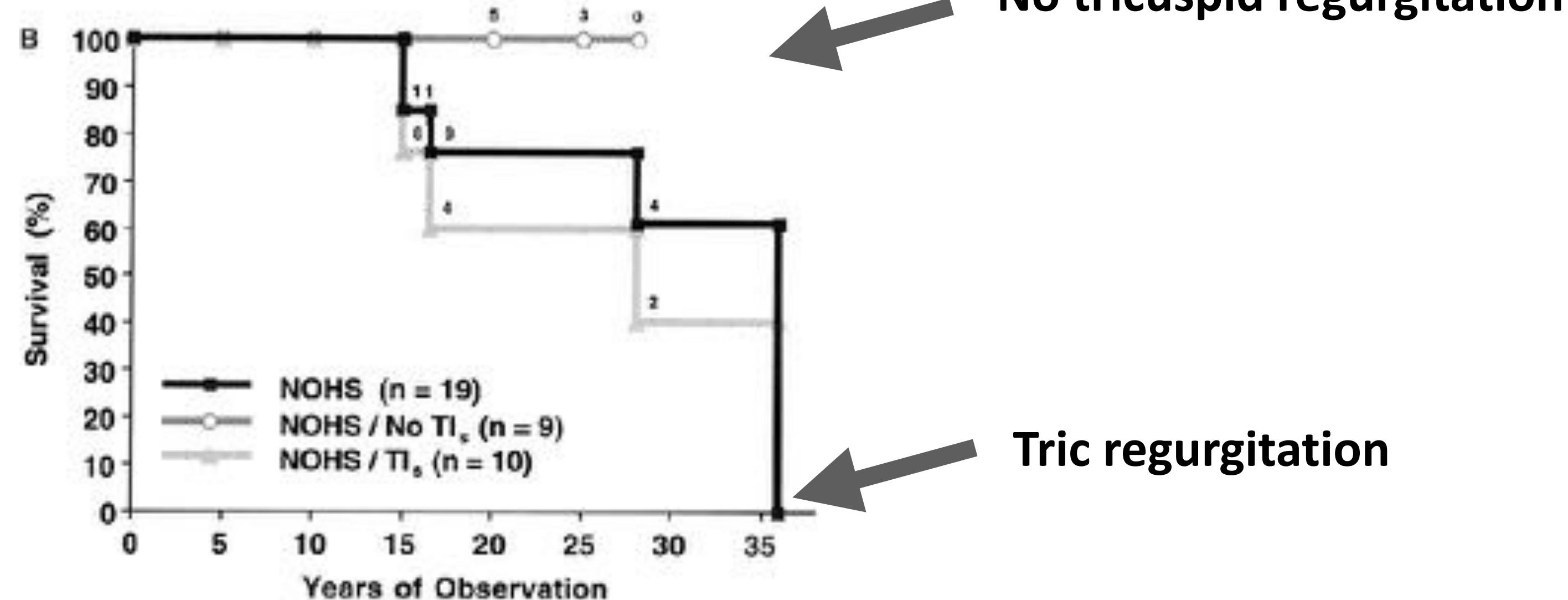
OHS



No tricuspid regurgitation

Tric regurgitation

No OHS

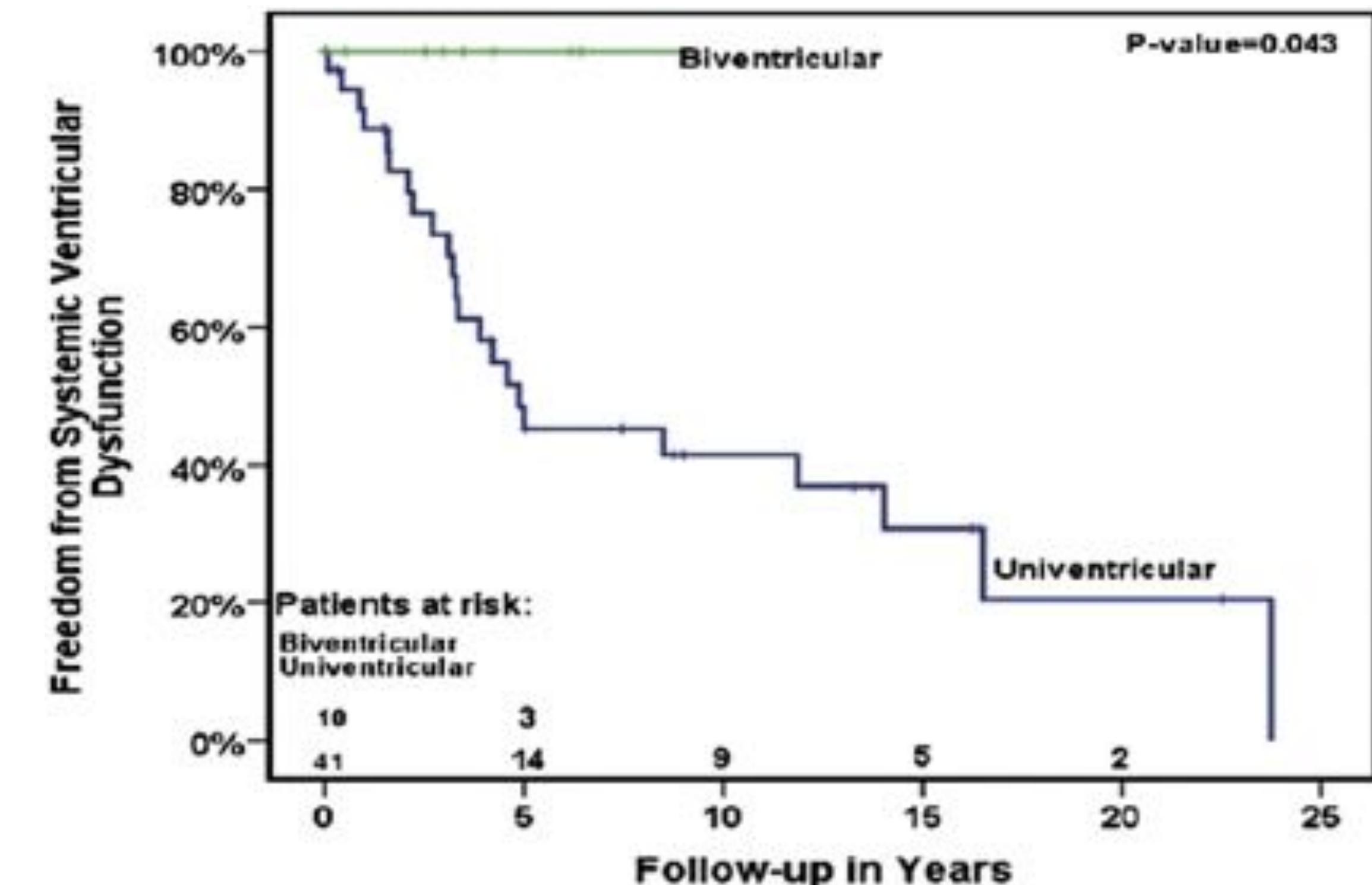


No tricuspid regurgitation

Tric regurgitation

Pacing for AV block

- Causes desynchronisation of ventricles
 - Worsening of tricuspid regurgitation
- Traumatic lesions of the mitral valve
- Need to stimulate both ventricles from the start
- Better do epicardial stimulation



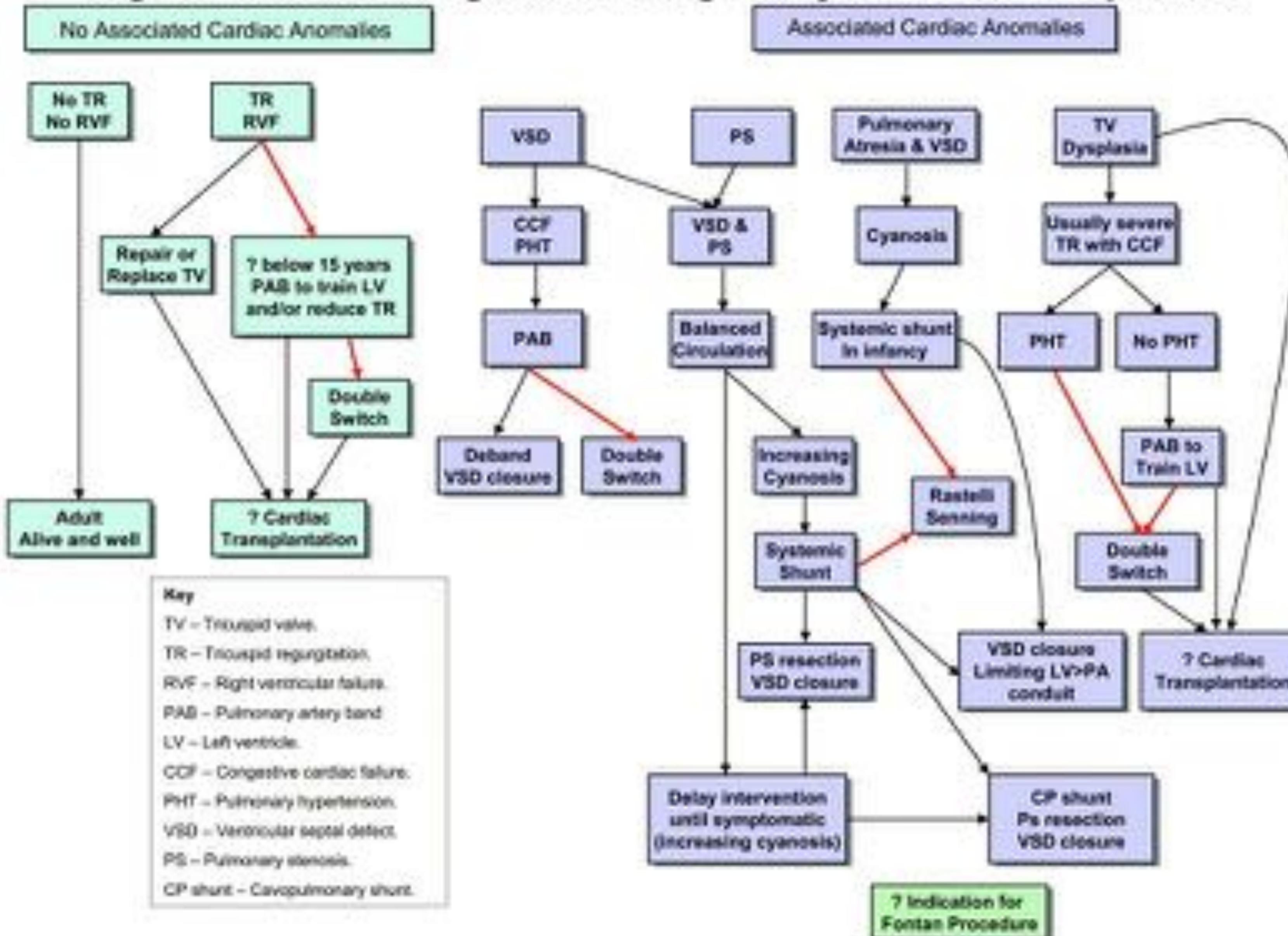
Double discordance

Surgical treatment

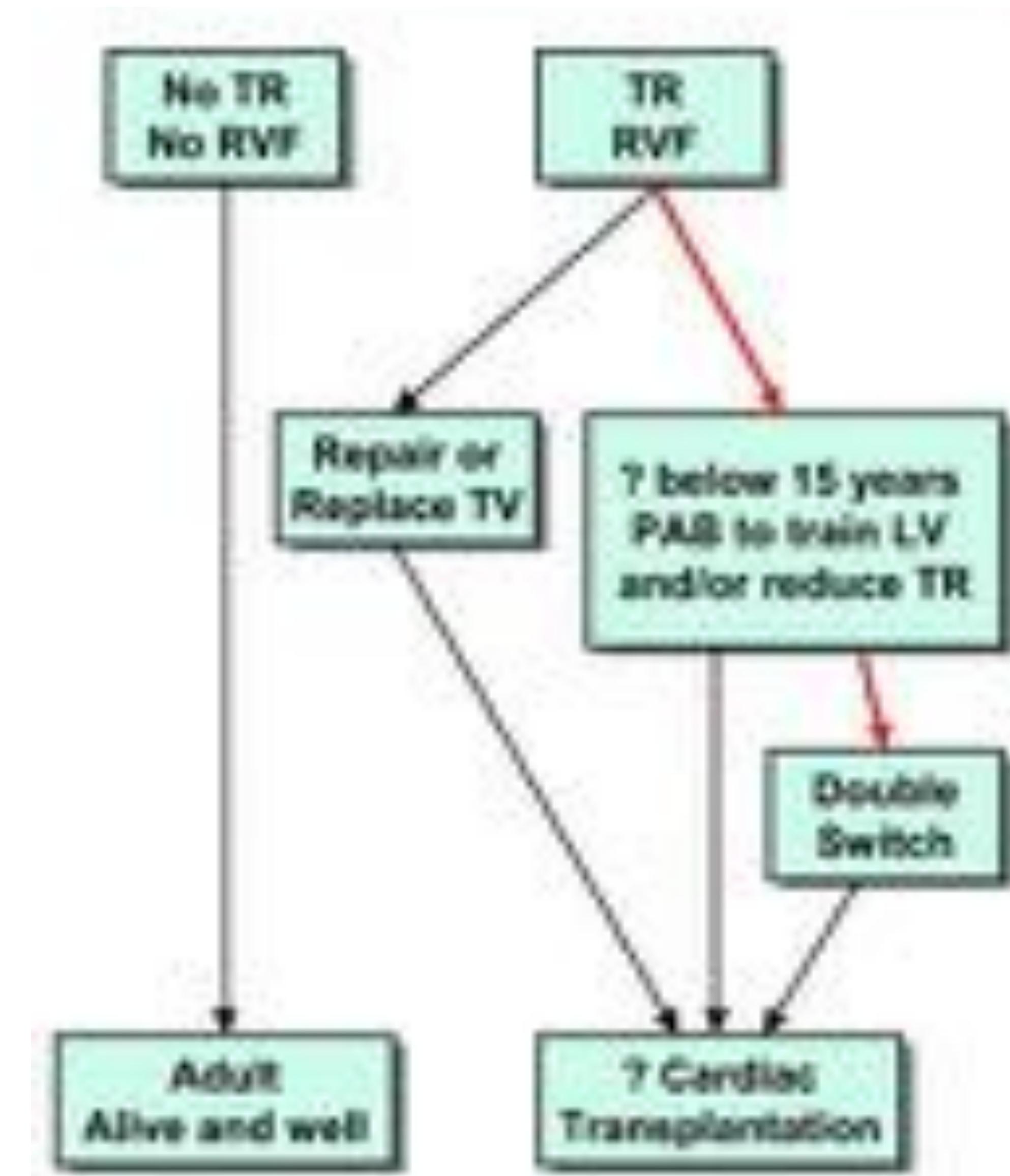
Therapeutic strategies

- Life long monitoring and intervention if needed
- Double switch or anatomical repair
- Conventional repair (treatment of associated anomalies : VSD, LVOTO-Pulmonary)
- Banding
 - retraining
 - preventive
- Fontan
- Transplantation

Algorithm for the management of Congenitally Corrected Transposition



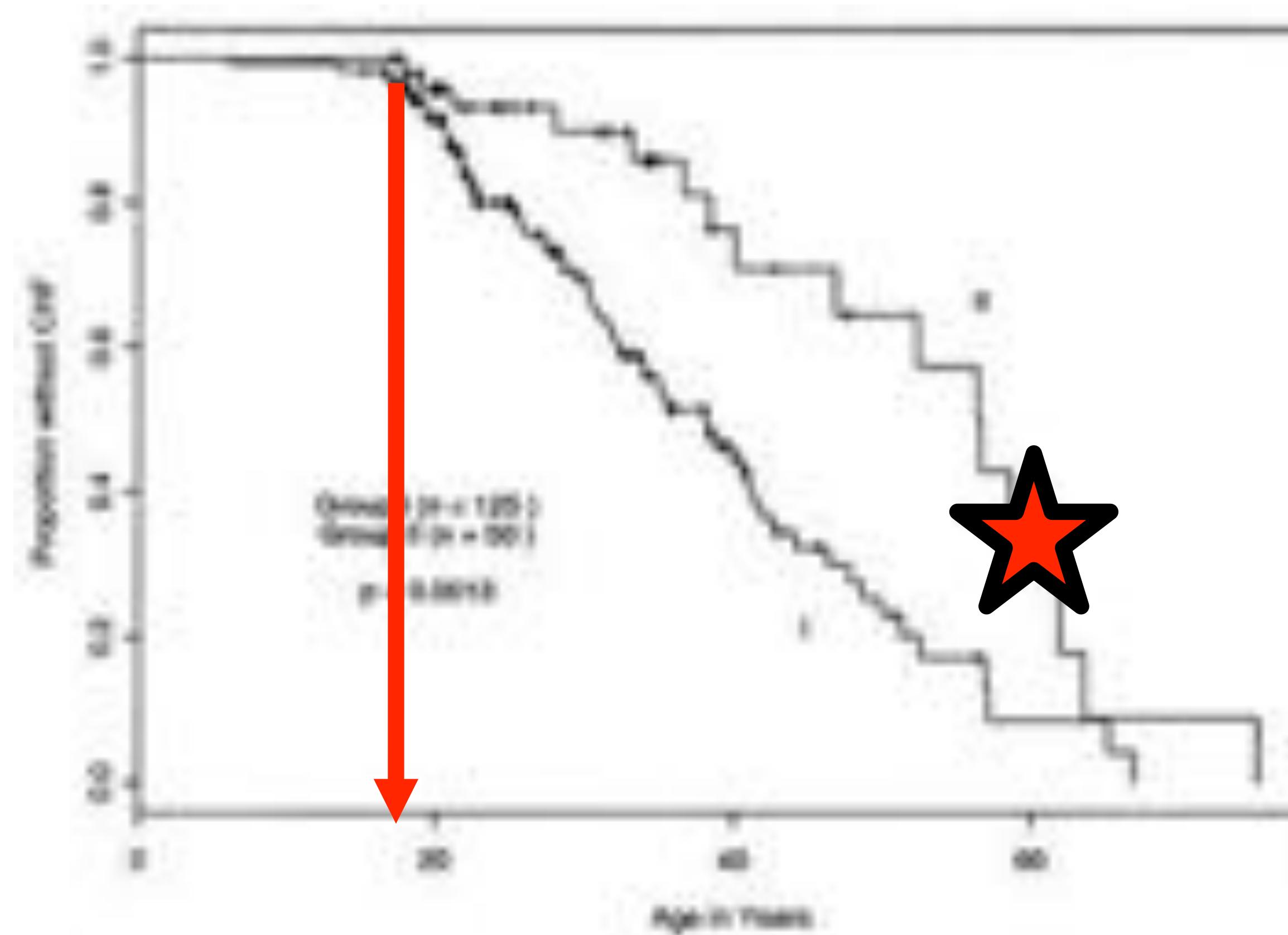
Life long monitoring strategy or conservative management Double discordance



Life long monitoring strategy

Double discordance - Survival in adulthood

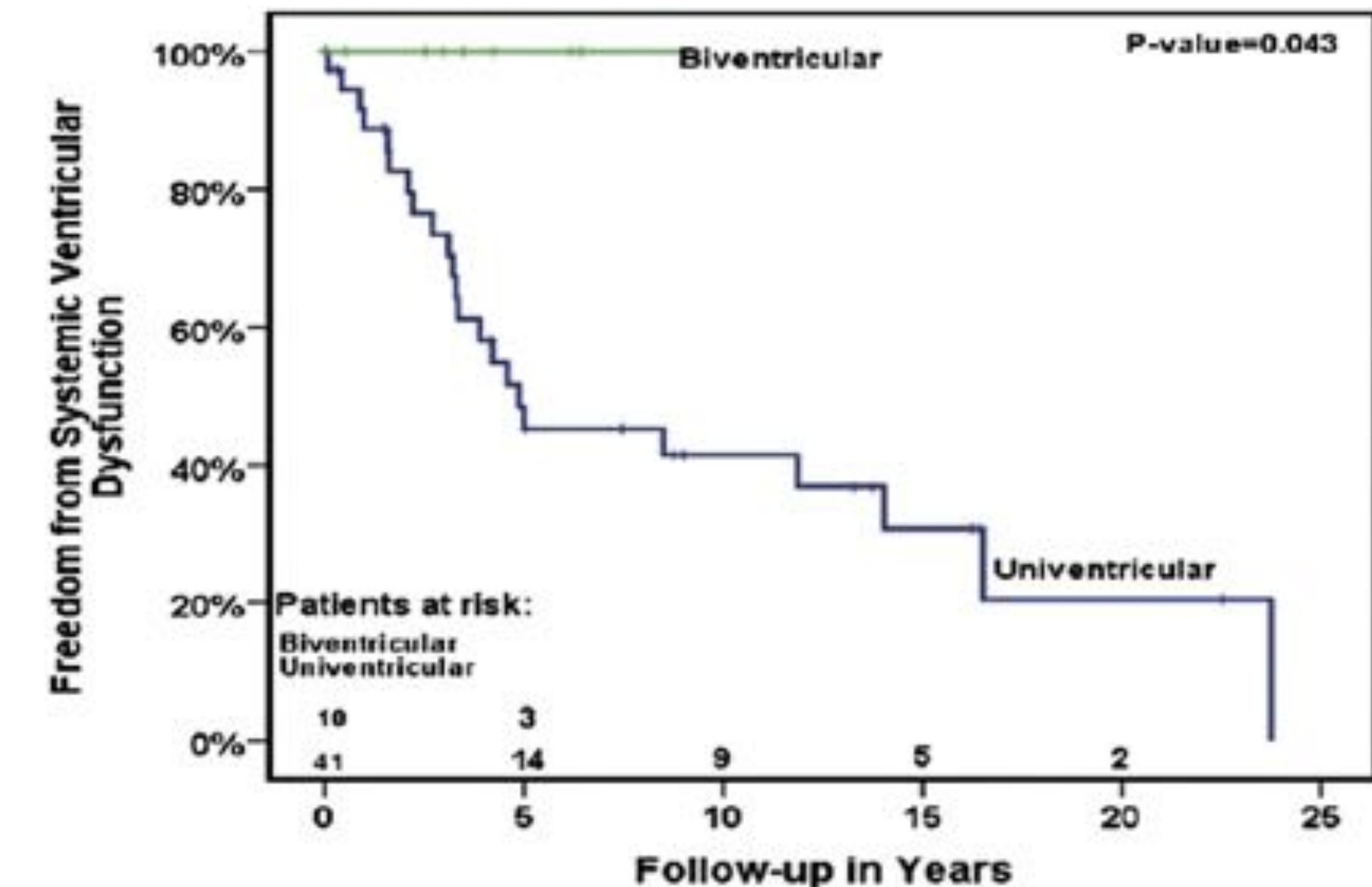
Survival without heart failure



- . Late outcomes are poorly understood
- . Late morbidity poorly understood
- . Do some groups perform better than others?

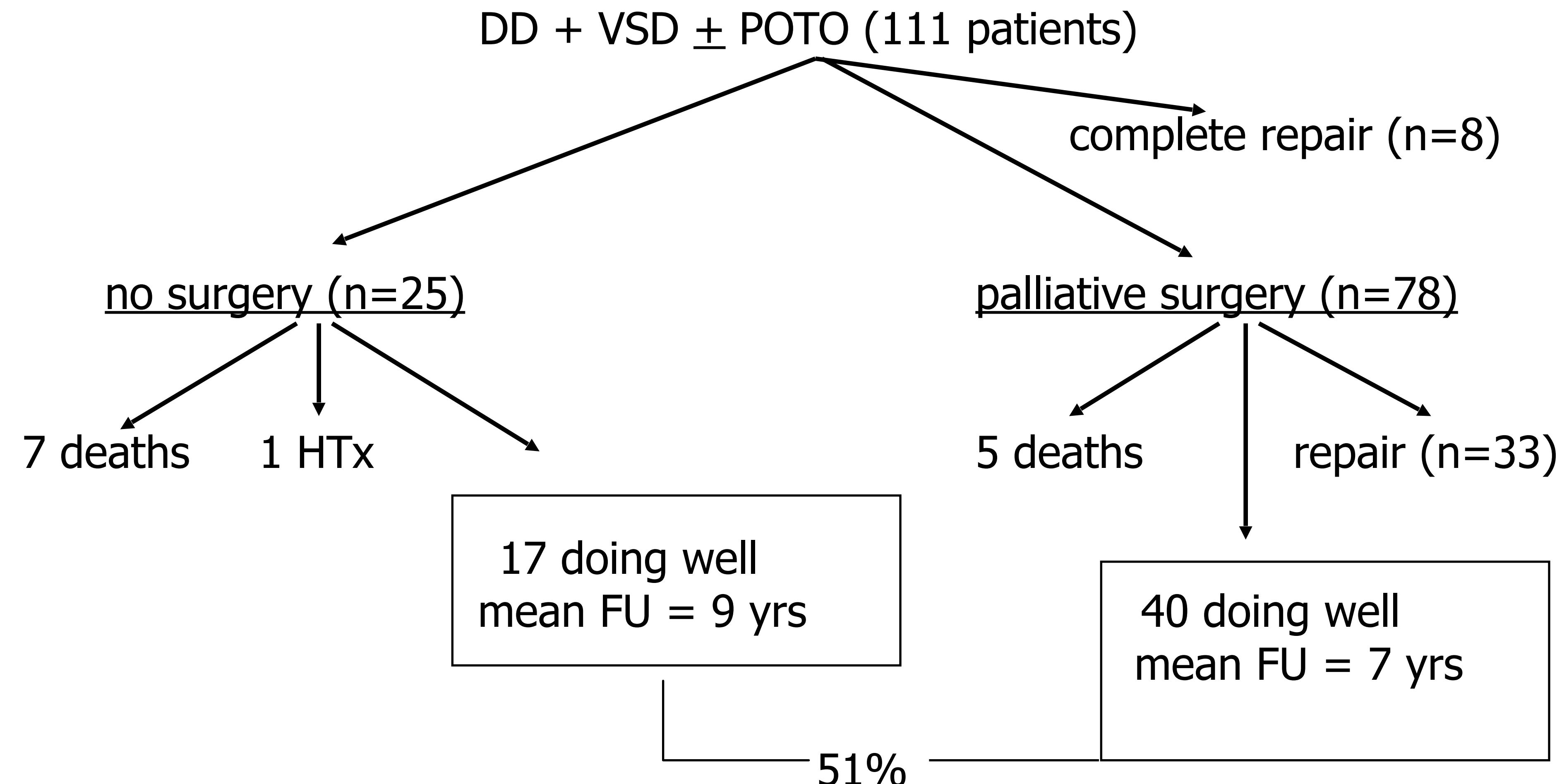
Pacing for AV block

- Causes desynchronisation of ventricles
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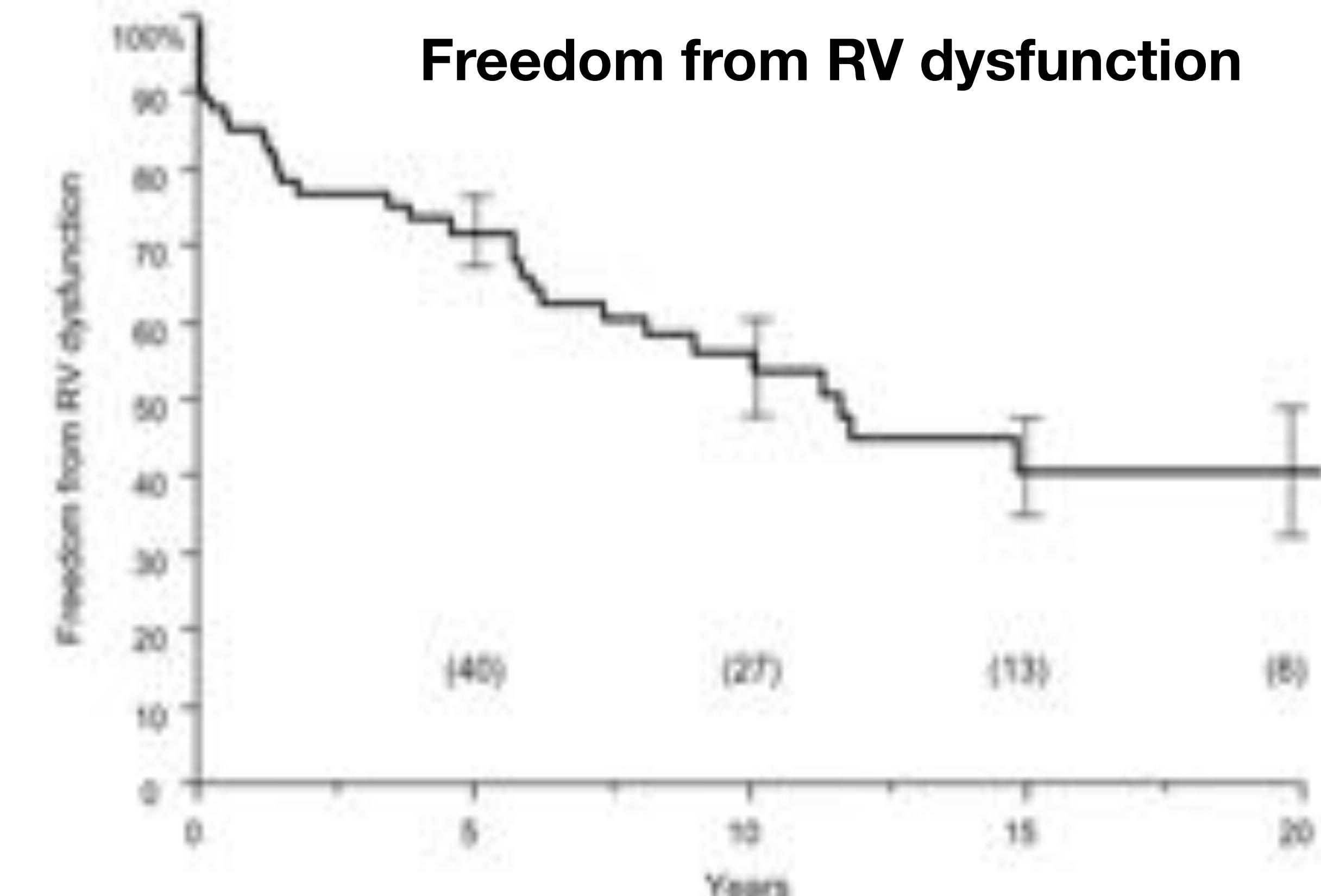
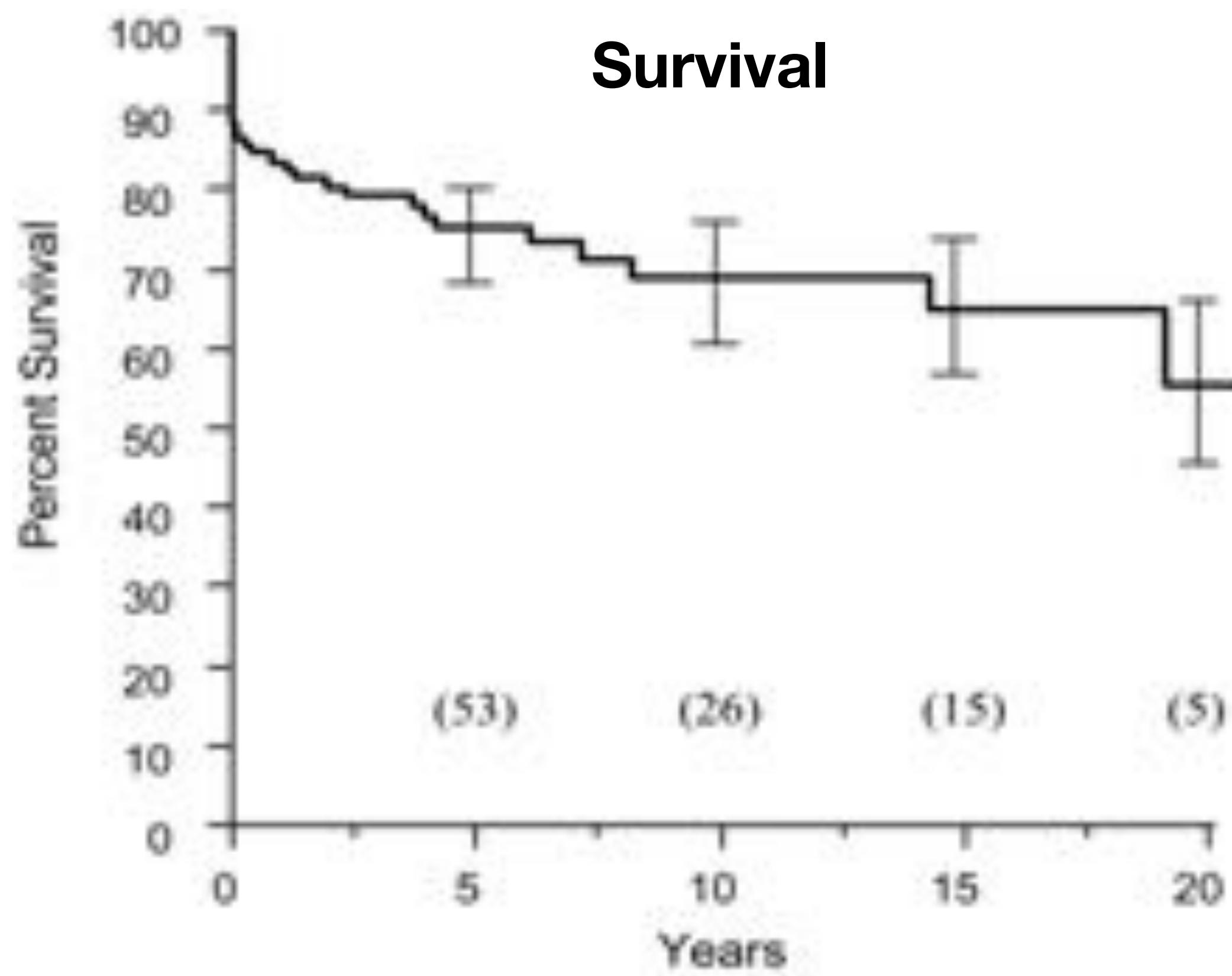
Repair/replace tricuspid valve

- Difficult decision
- Right ventricular dysfunction is usually associated
- ...and does not always improve with tricuspid valve replacement
- Surgical mortality is high 15%
- Mechanical valve is the « best » option



Conventional repair

RV still systemic

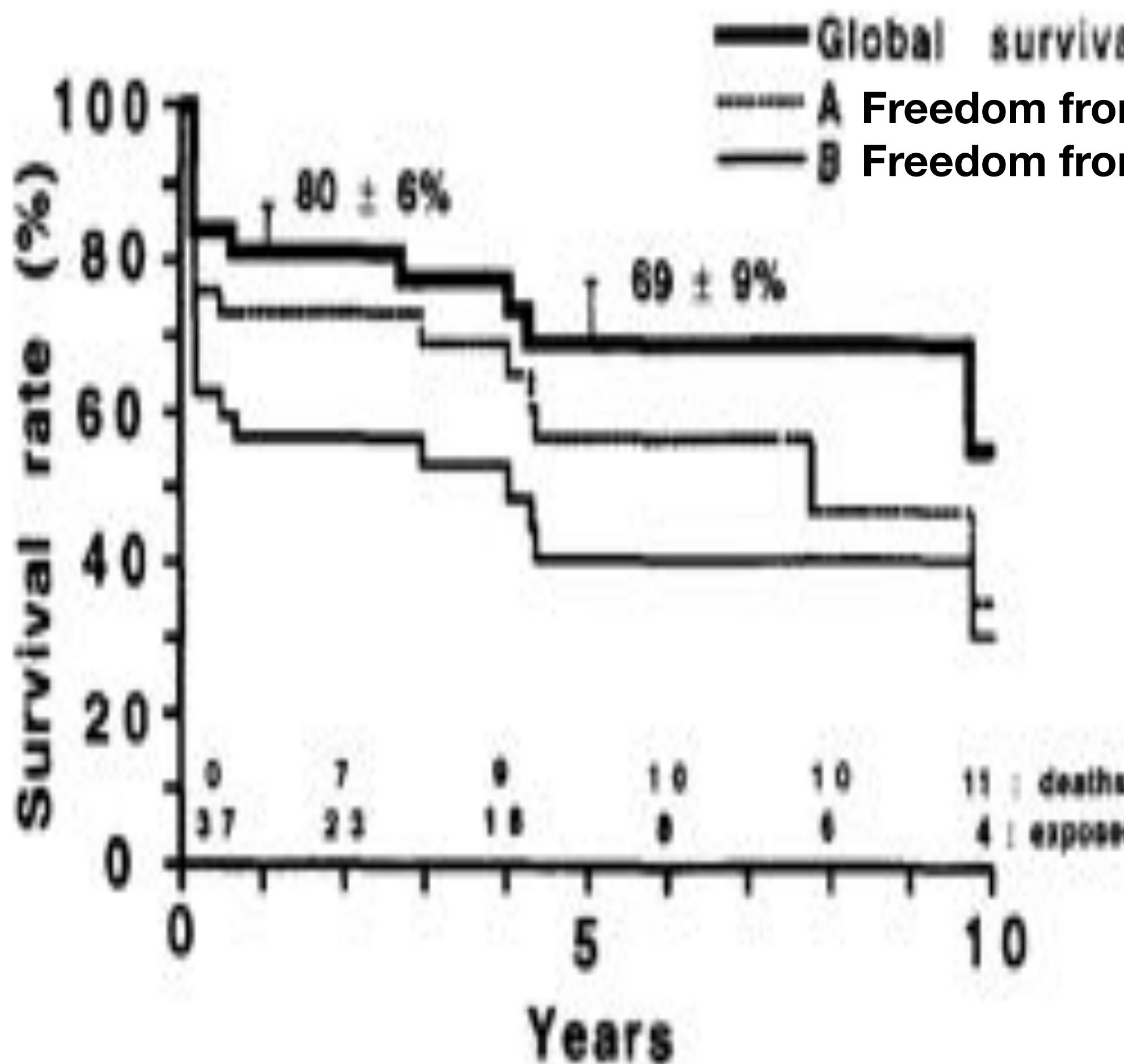


Hraska V et al. Long-term outcome of surgically treated patients with corrected transposition of the great arteries. J Thorac Cardiovasc Surg 2005;129:182-191

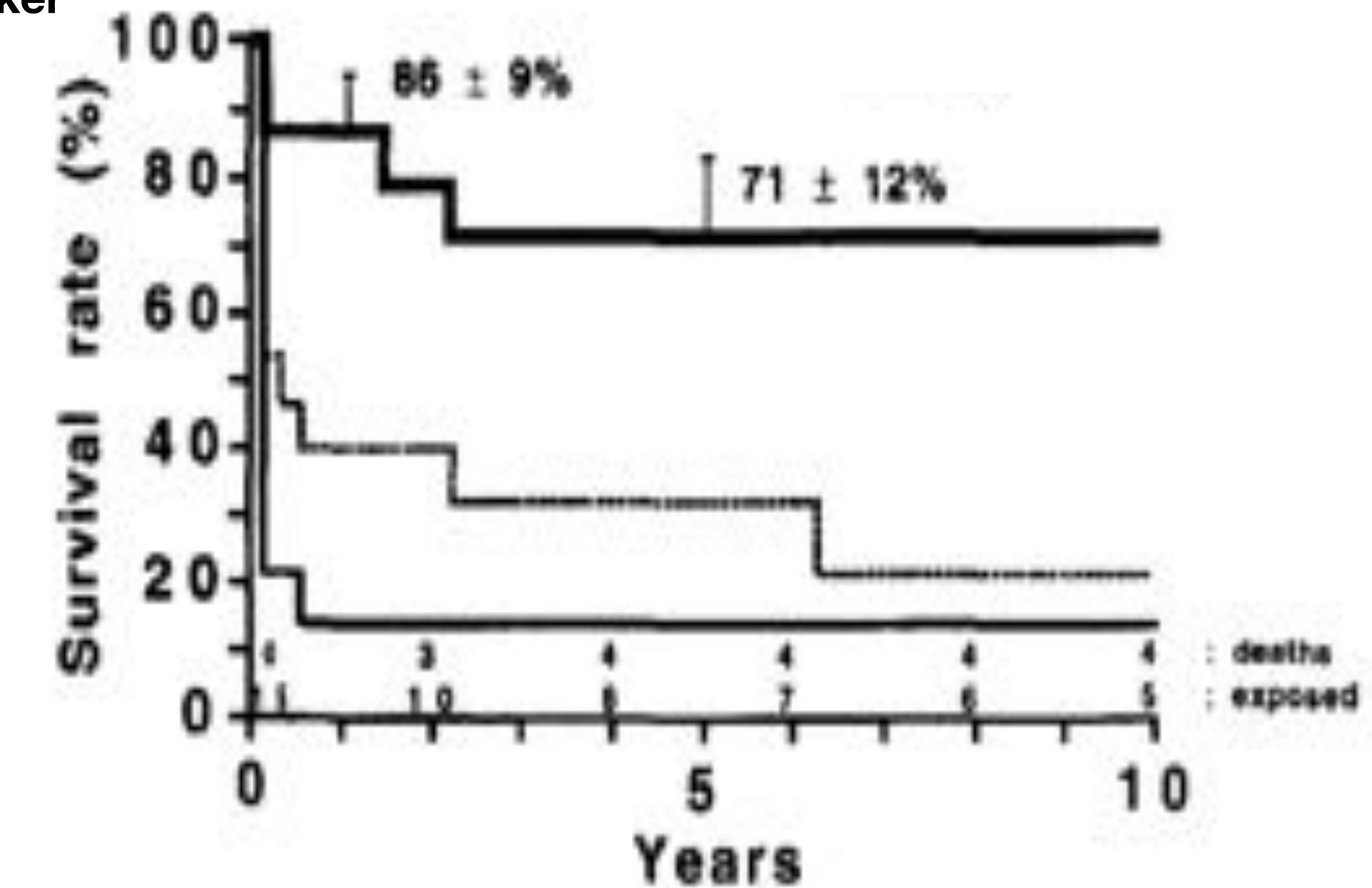
Conventional repair

RV still systemic

DD+VSD+LVOTO



DD+VSD

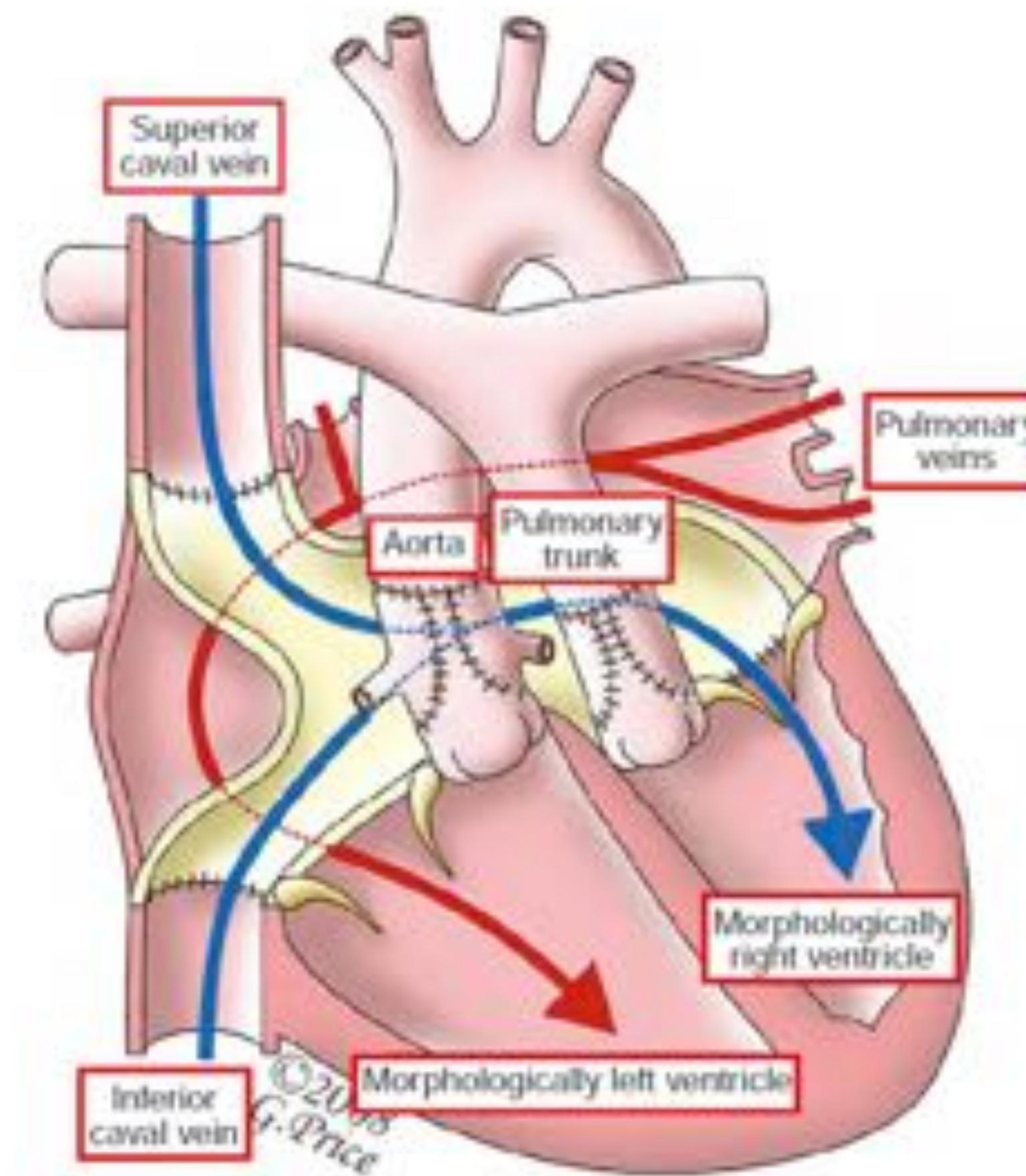


Conventional repair

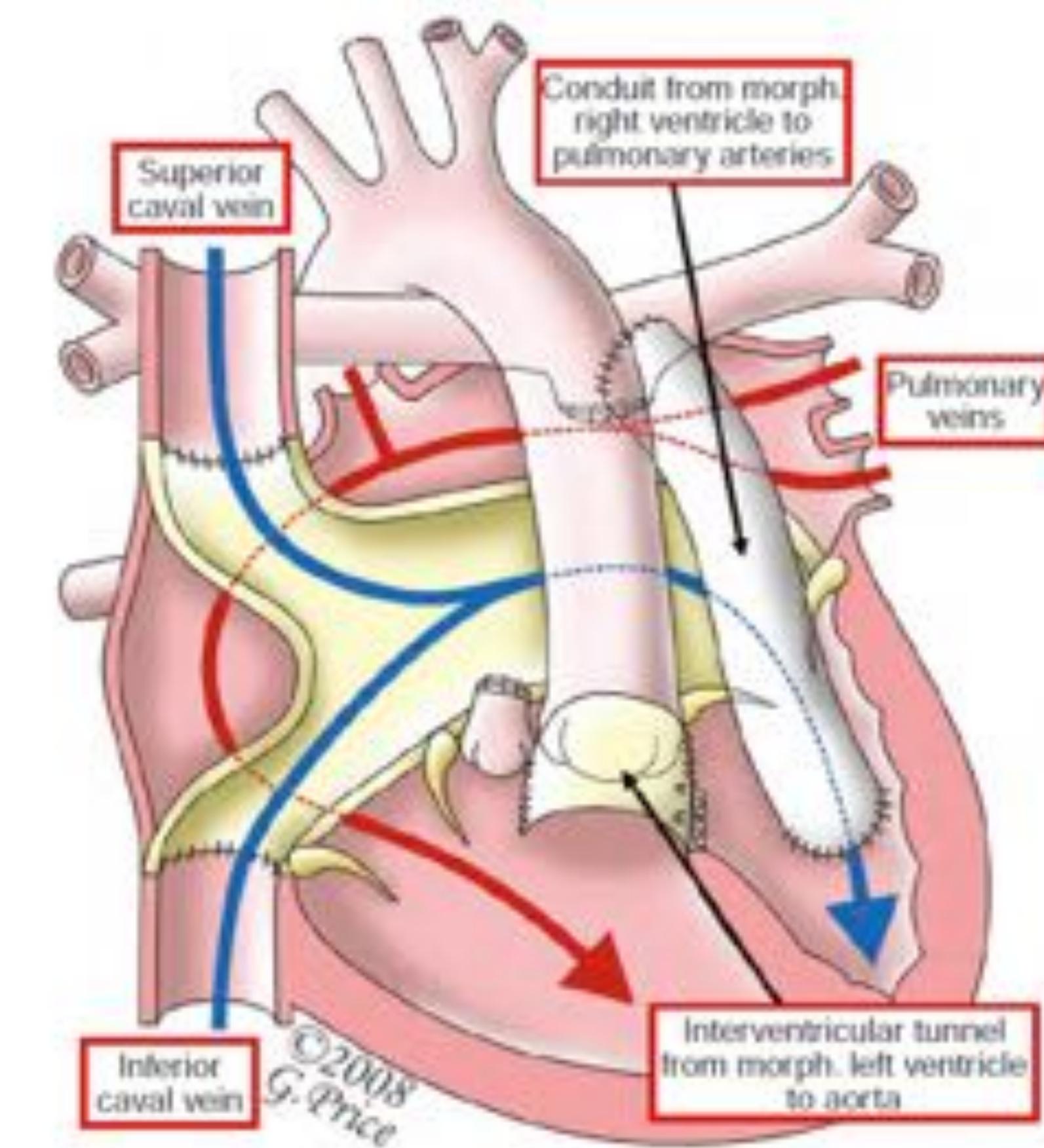
RV still systemic

- provides disappointing results in ccTGA + VSD
 - high incidence of TVR and RV dysfunction
 - should be avoided
- may be an option in ccTGA + VSD + POTO
 - contra-indication to anatomic repair
 - unfavorable anatomy for anatomic repair
 - with incomplete relief of POTO
(elevated LV pressure and flat septum)

Principle of anatomic repair



Double-Switch



Senning-Rastelli

Anatomic repair

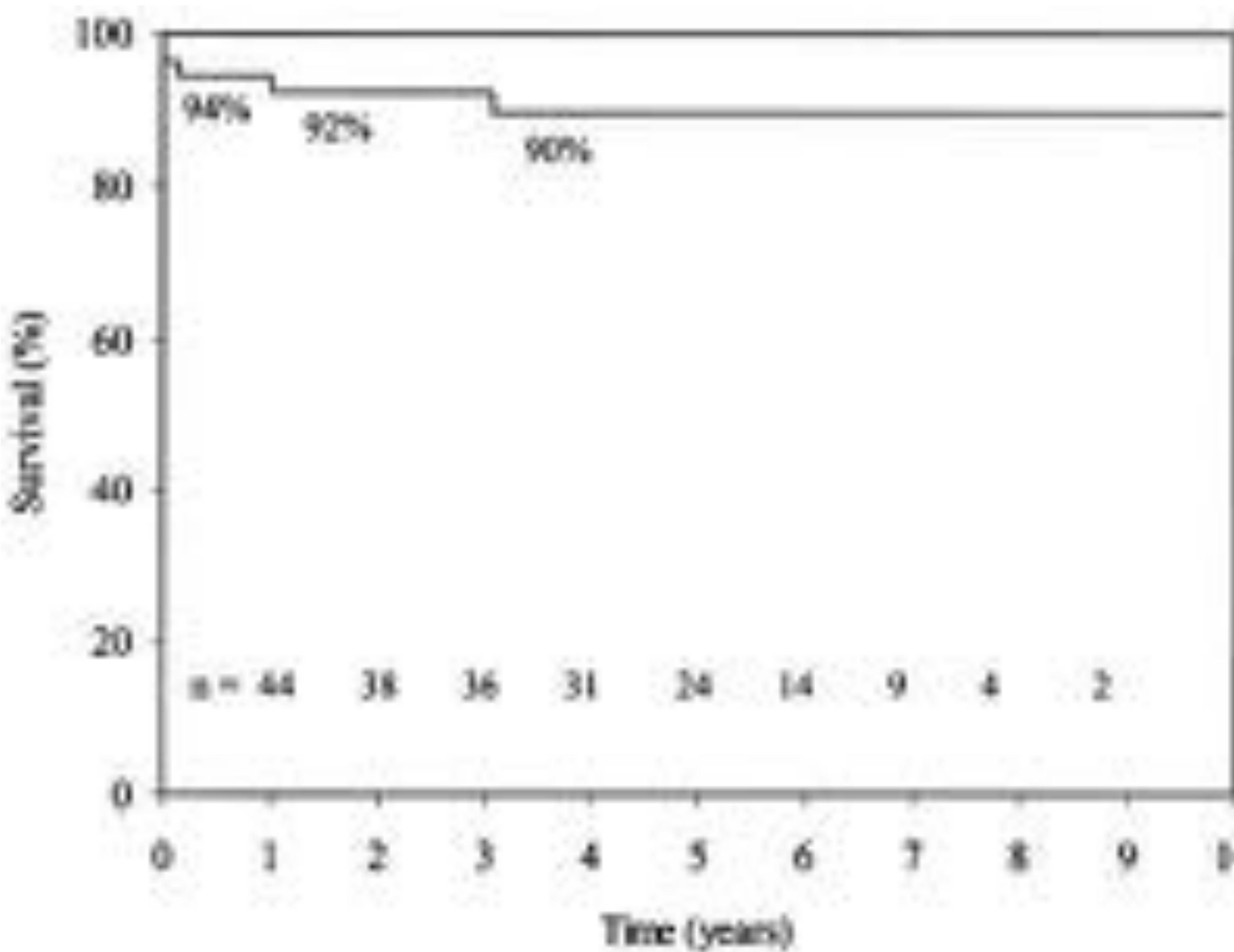
Pre-requisites

- **Mitral valve**
 - normal
 - easy to repair (mitral cleft)
- **Left ventricle**
 - normal in size and function
 - ready to sustain systemic workload
(LV retraining may be necessary)

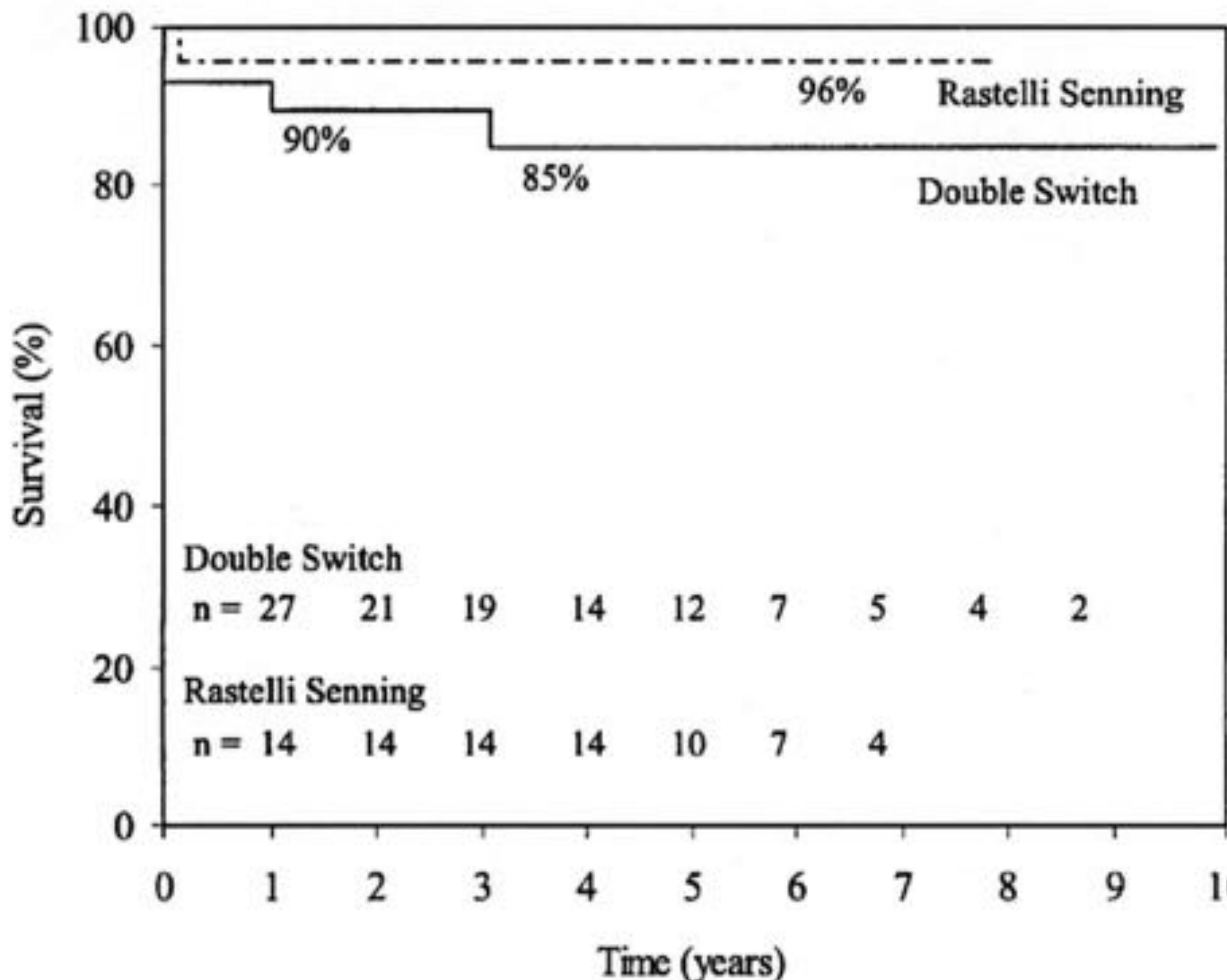
Double switch or Senning-Rastelli in ccTGA

- Prophylactic : Yes we can !
- As a strategy if non restrictive VSD or prophylactic PA banding
 - With always good LV
- If needed in case of RV dysfunction or tricuspid regurgitation
 - Failure of LV retraining : 20%
 - Success of LV retraining: 50%

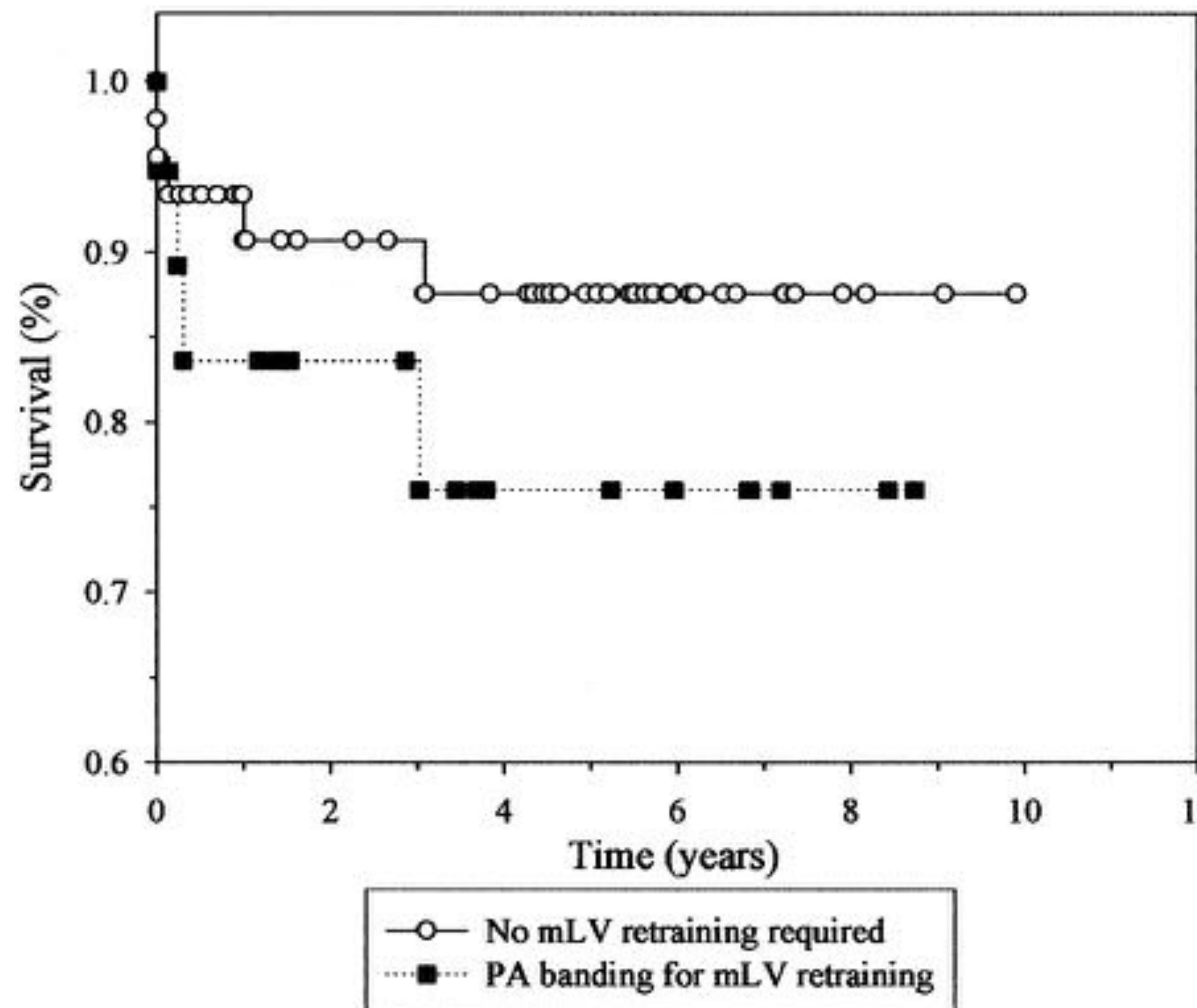
Survival after double switch



Survival after double switch or Rastelli-Senning



Survival after double switch with or without banding for LV retraining

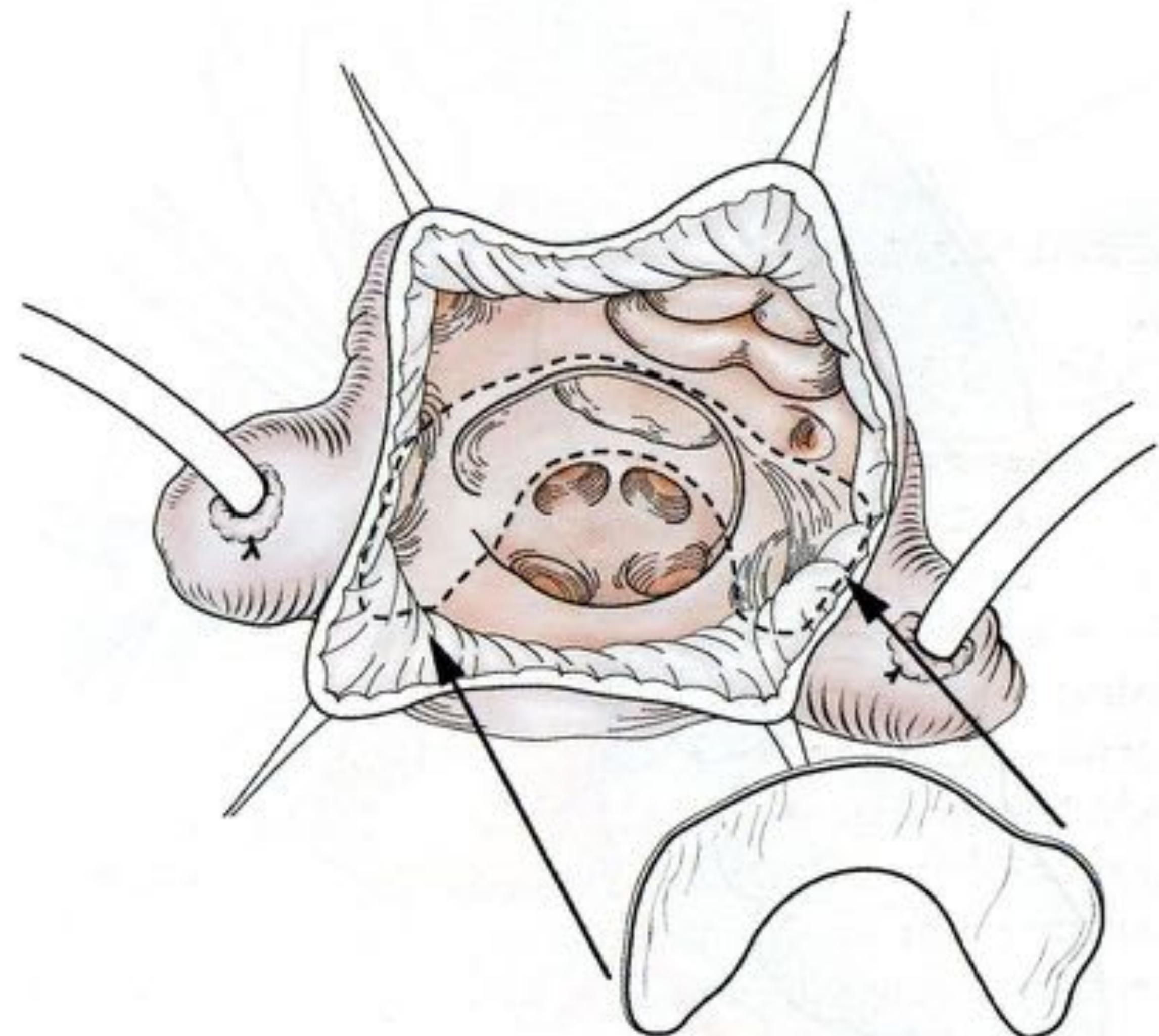


Anatomic repair

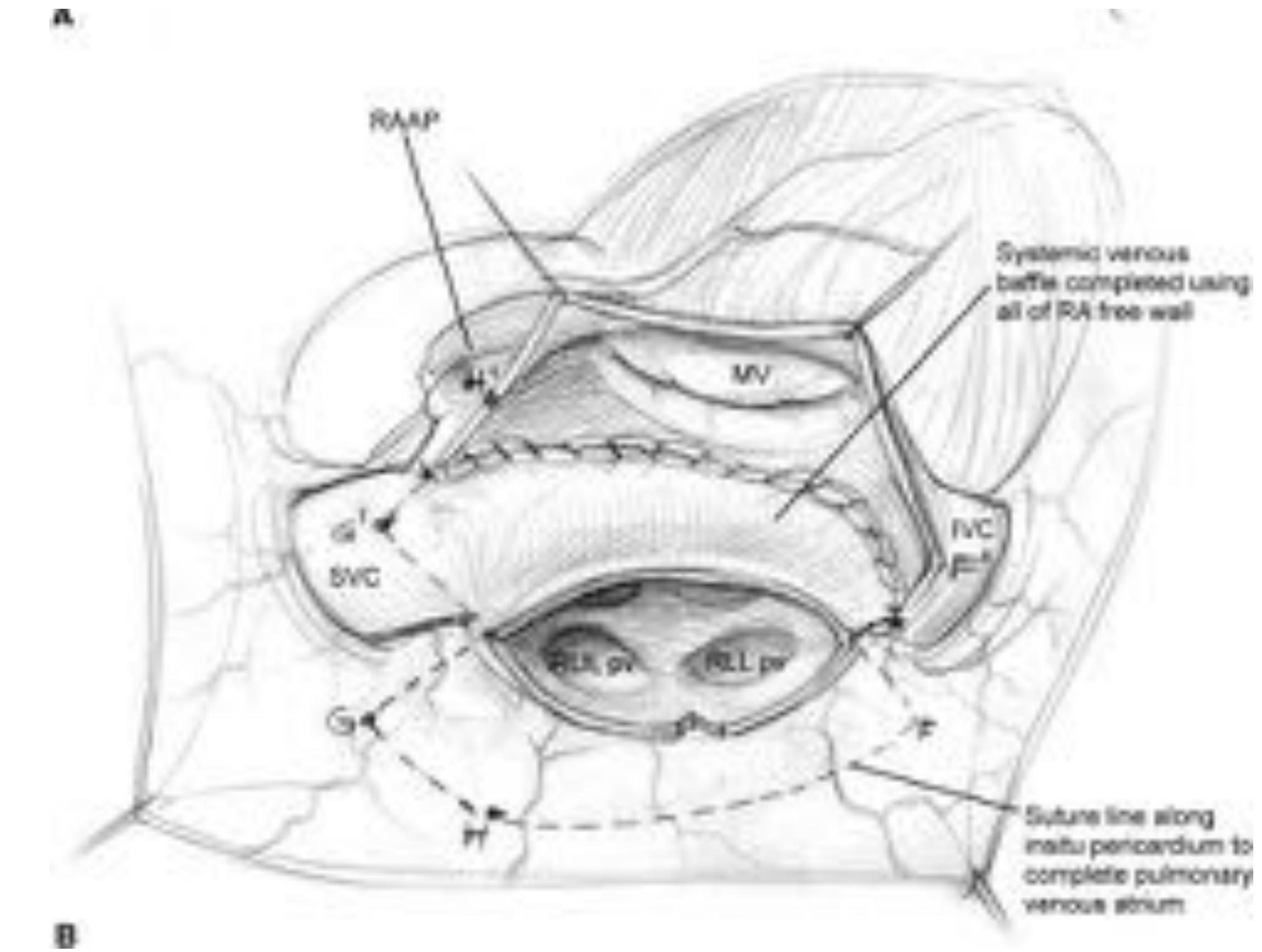
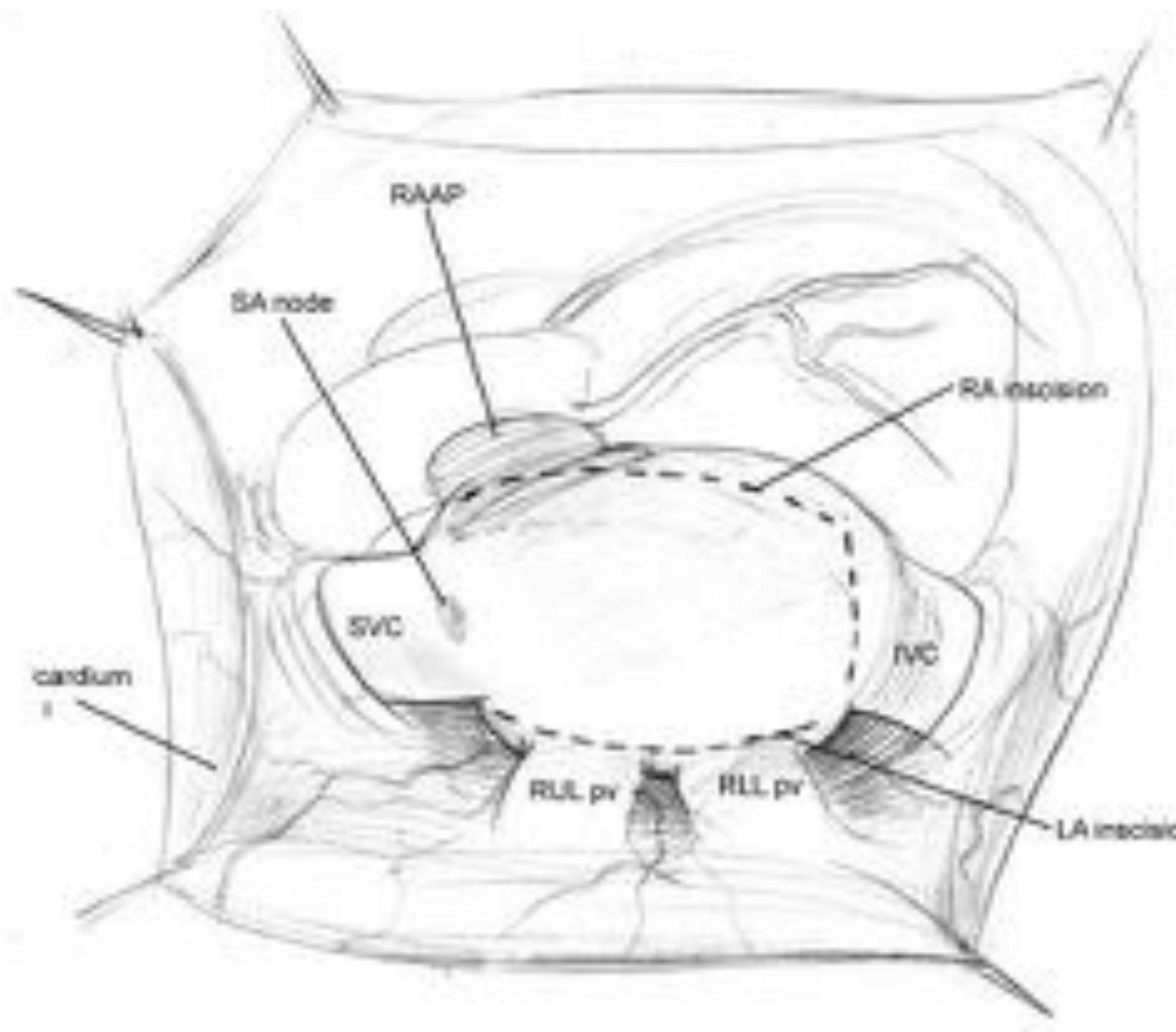
The atrial switch in ccTGA

- **Surgical factors**
 - cardiac position
 - size of atria
 - juxtaposition of atrial appendages
 - position of conduction tissue
- **Surgical choices**
 - Mustard procedure
 - Senning procedure
 - hemi-Mustard + cavopulmonary shunt
 - extracardiac atrial switch

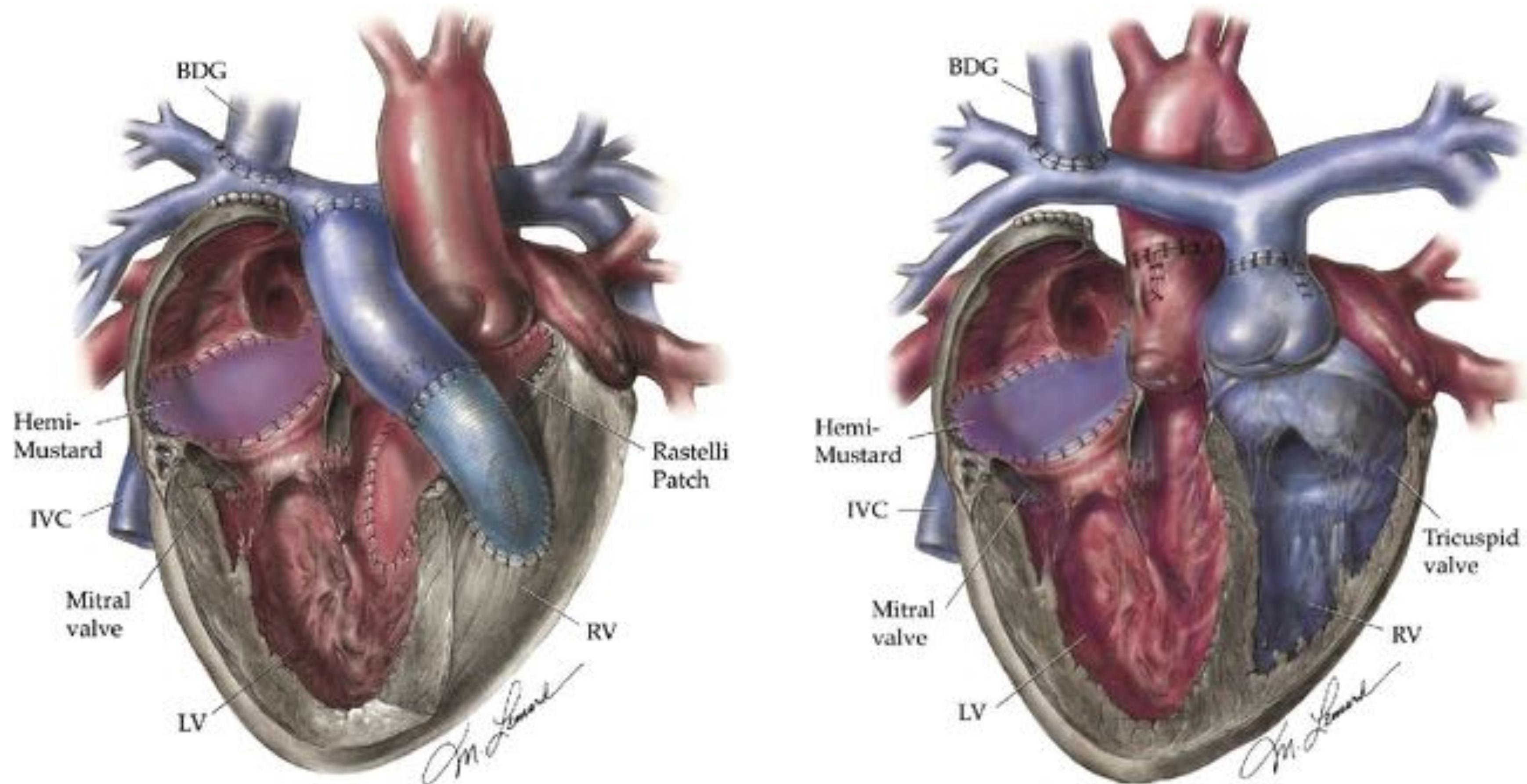
Mustard procedure



Senning operation (Shumaker modification)



Hemi-Mustard + cavopulmonary shunt



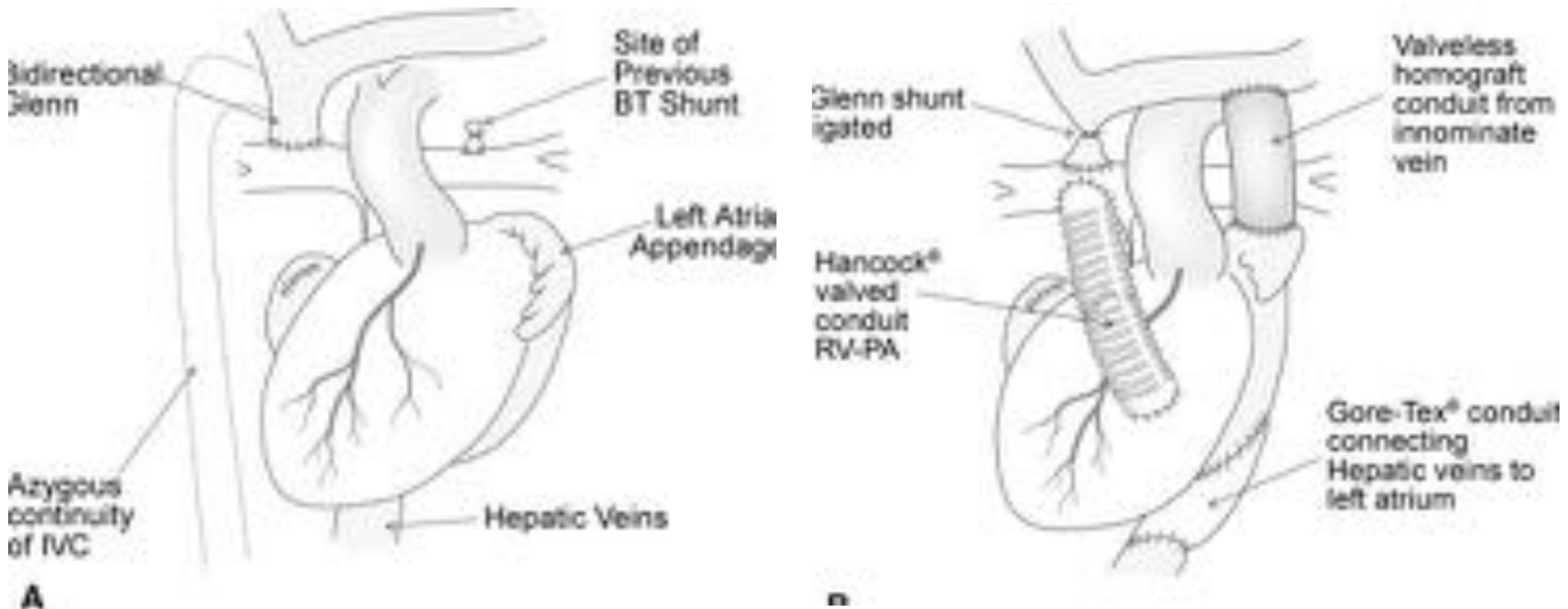
n= 48.

One early death and no late deaths.

13% have significant impaired LV function - All were in the Double Switch group

Reoperation rate not different between Rastelli/Mustard and Double Switch

Extracardiac atrial switch

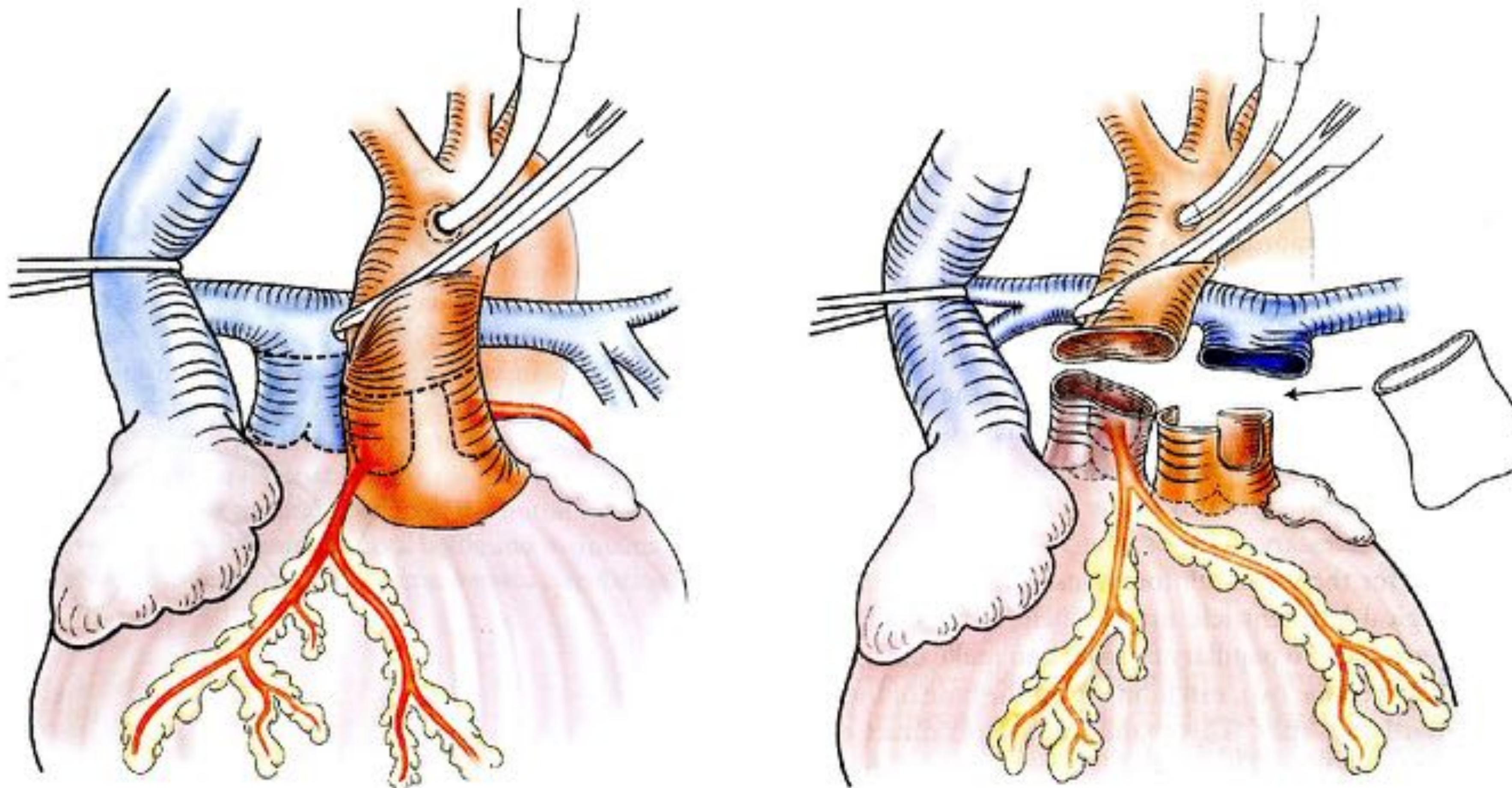


Anatomic repair

The arterial switch in ccTGA

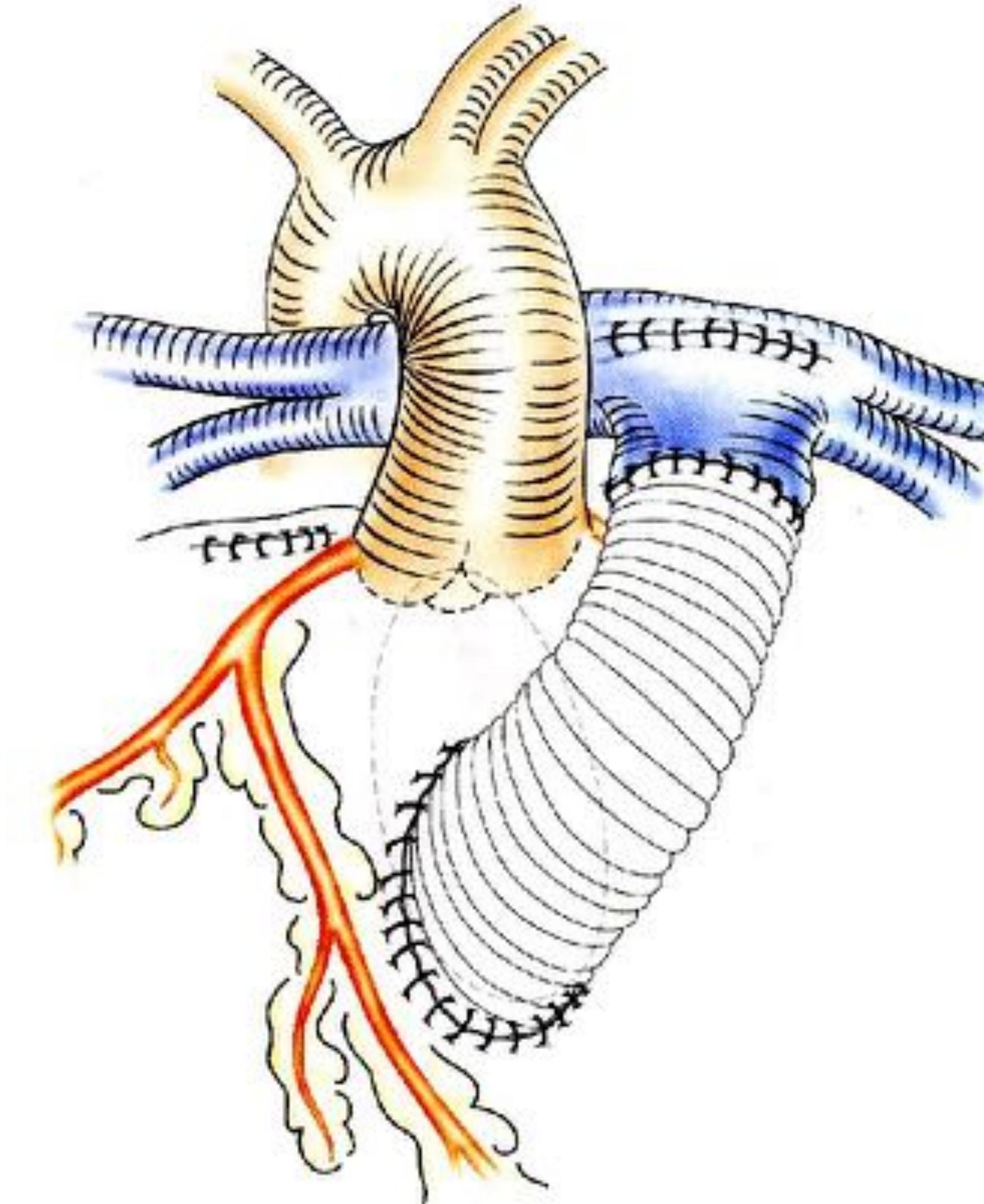
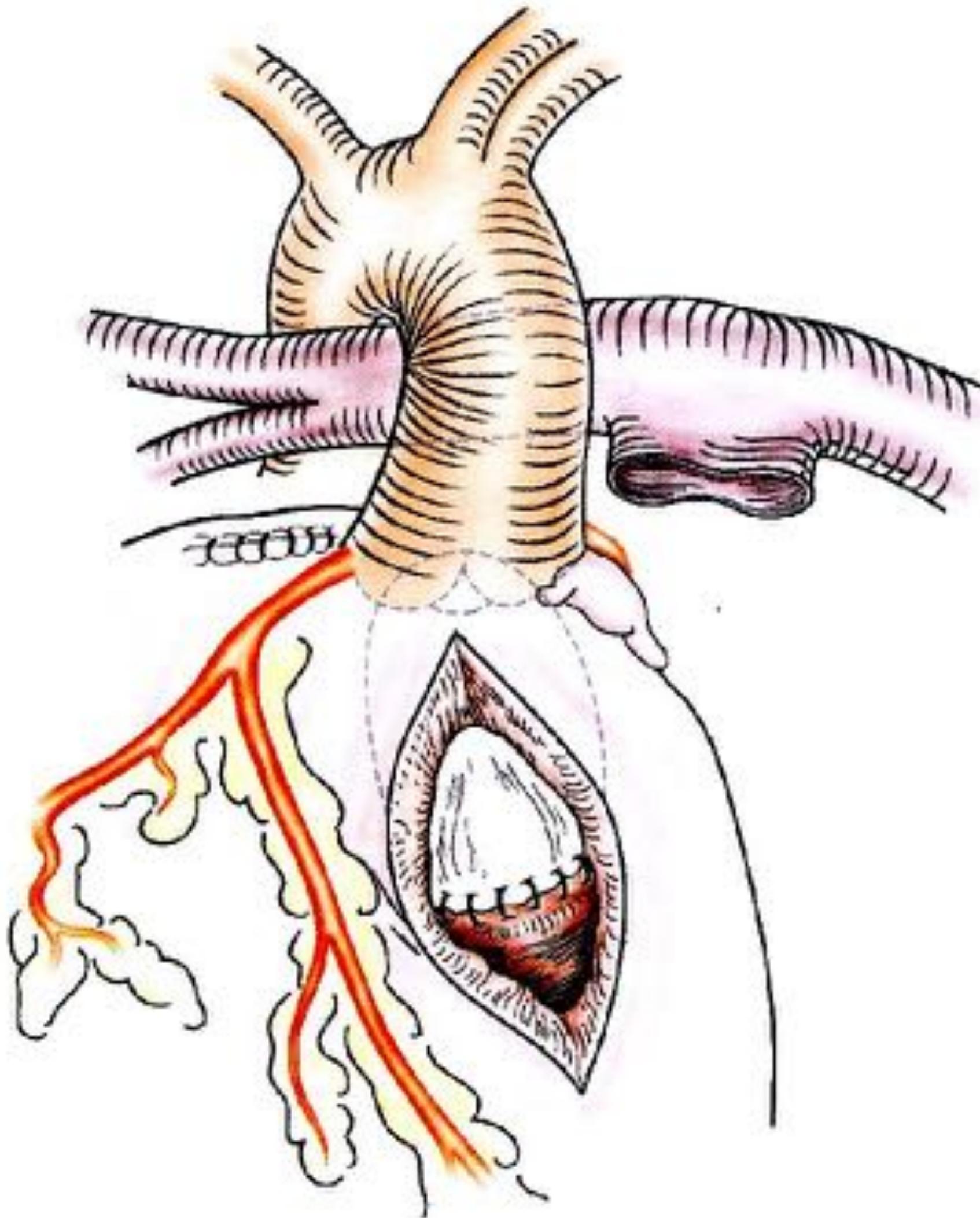
- **Arterial switch**
 - normal pulmonary outflow tract
 - resectable subpulmonary stenosis
- **Associated pulmonary stenosis**
 - LV – aorta rerouting (Rastelli)
 - aortic translocation (Bex-Nikaidoh)

Anatomic repair Arterial switch operation in ccTGA

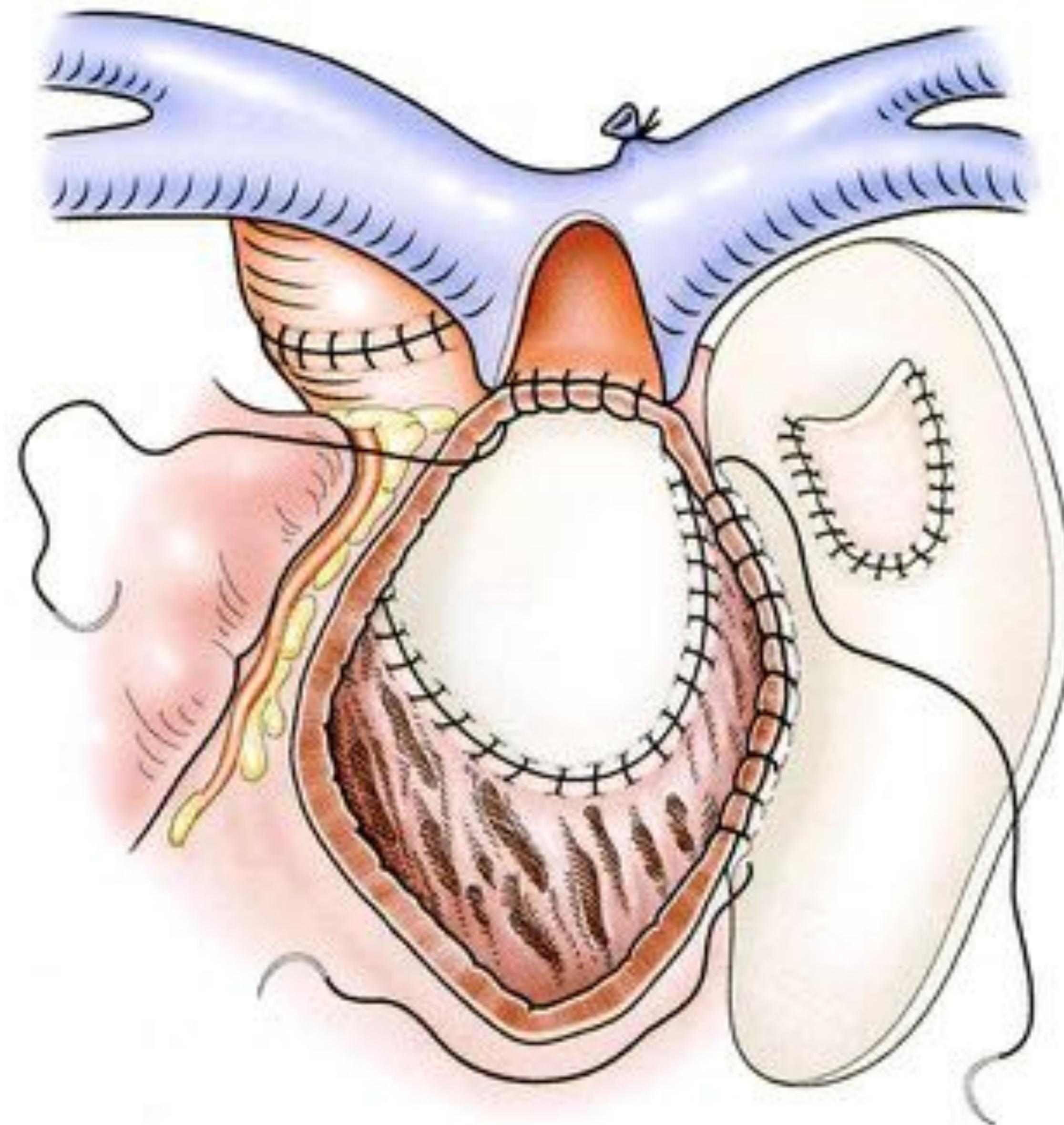


Anatomic repair

Rastelli – type repair



REV reconstruction of the RVOT



Anatomic repair Potential complications

- **Related to atrial switch**
 - arrhythmias
 - systemic or pulmonary venous obstruction
- **Related to arterial switch**
 - coronary complications
 - aortic insufficiency
 - pulmonary stenosis
- **Related to Rastelli procedure**
 - obstruction of RV-PA conduit
 - subaortic stenosis
- **Related to ccTGA**
 - tricuspid regurgitation
 - LV function (preliminary banding)

Pulmonary artery banding in ccTGA

- Mandatory
 - in ccTGA with VSD
- Treatment
 - of tricuspid regurgitation to restore septal geometry and reduce tricuspid regurgitation volume
- Prophylactic or as destination therapy
 - in all neonates with ccTGA



Thank you