Cyanotic Congenital Heart Disease

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Cardiology

UTHSCSA and STVHCS

Recommended References

- Perloff, JK. Clinical Recognition of Congenital Heart Disease. 5th ed. 2003.
- Mavroudis C et al. Pediatric Cardiac Surgery. 3rd ed. 2003.
- Allen HD et al. Moss and Adams' Heart Disease in Infants, Children, and Adolescents. 6th ed. 2001.
- Braunwald E et al. Heart Disease; a Textbook of Cardiovascular Medicine. 7th ed. 2005.
- www.cachnet.org (Canadian Adult Congenital Heart Network)
- www.achd-library.com (The Nevil Thomas Adult Congenital Heart Library)

Outline

- Epidemiology and Pathophysiologic considerations
- Specific lesions
  - Tetralogy of Fallot
  - Transposition of the Great Arteries
  - Truncus Arteriosus
  - Tricuspid Atresia
  - Total Anomalous Pulmonary Venous Return

Abnormal Developmental Mechanisms

- Conus and great vessel development
- Intracardiac blood flow
  - Valve stenosis, atresia
  - ASD, VSD
- Cell death abnormality
- Extracellular matrix
- Abnormal targeted growth
- Abnormal situs and looping

TABLE 5.3. PATHOGENETIC CLASSIFICATION OF SOME CONGENITAL CARDIOVASCULAR MALFORMATIONS BASED ON COMMON DEVELOPMENTAL MECHANISM RATHER THAN ANATOMIC DETECT

- Ectomesenchymal tissue migration abnormalities
  - Conotruncal septation defects
    - Subarterial, type I ventricular septal defect
    - Double-outlet right ventricle
    - Tetralogy of Fallot
    - Pulmonary atresia with ventricular septal defect
    - Aorticopulmonary window
    - Truncus arteriosus communis
  - Abnormal conotrucal cushion position
    - Transposition of the great arteries (–d)
  - Branchial arch defects
    - Interrupted aortic arch type B
    - Double aortic arch
    - Right aortic arch with mirror-image branching
- Abnormal intracardiac blood flow
  - Perimembranous ventricular septal defect
  - Left heart defects
    - Biscuspid aortic valve
    - Aortic valve stenosis
    - Coarctation of the aorta
    - Interrupted aortic arch type A
    - Hypoplastic left heart, aortic atresia:mitral atresia
  - Right heart defects
    - Biscuspid pulmonary valve
    - Secundum atrial septal defect
    - Pulmonary valve stenosis
    - Pulmonary valve atresia with intact ventricular septum
- Cell death abnormalities
  - Muscular ventricular septal defect
  - Ebstein's malformation of the tricuspid valve
- Extracellular matrix abnormalities
  - Endocardial cushion defects
    - Ostium premium atrial septal defect
    - Type III, inflow ventricular septal defect
    - Atrioventricular septal defect
  - Dysplastic pulmonary or aortic valve
- Abnormal targeted growth
  - Anomalous pulmonary venous return
- Abnormal situs and looping
  - Heterotaxia, L-loop

?PDA

Moss and Adams, 2001, p. 68

TABLE 5.5. PREVALENCE OF SELECTED CONGENITAL CARDIOVASCULAR MALFORMATIONS PER 10,000 LIVE BIRTHS FROM CASES REGISTERED IN THE BALTIMORE–WASHINGTON INFANT STUDY, 1981–1989

	Defect	Prevalence per 10,000 live births					
7	*Transposition of the great arteries	7	*	2.64	T	8	4
	*Truncus arteriosus		*	0.69	T	9	2
	*Double-outlet right ventricle		*	0.49	*		
8	*Tetralogy of Fallot	8	*	2.60	T	7	6
6	AV septal defect	6		3.27			
10	Trisomy 21	10		2.32			
	Euploid			0.97			
	*Total anomalous pulmonary venous return		*	0.66	T		
5	*Tricuspid valve atresia, normal great vessels	5	*	3.6	T	10	1
	Ebstein's anomaly of the tricuspid valve			0.52			
11	*Hypoplastic left heart syndrome	11	*	1.78[a]	*		
4	Pulmonary valve stenosis	4		3.78[b]		4	7
2	*Pulmonary valve atresia, intact IVS	?2	*	5.8	*		
	Aortic valve stenosis, bicuspid aortic valve			0.81/0.74		6	6
13	Coarctation of the aorta	13		1.39		5	7
	Ventricular septal defect			15.57		1	30
1	Perimembranous	1		9.87			
3	Muscular	3		4.7[b]			
9	Atrial septal defect (isolated secundum type)	9		2.35		2	10
	Patent ductus arteriosus			0.88		3	10
12	*Laterality and looping including LTGA	12	*	1.44	*		

Moss and Adams, 2001. P. 69; Braunwald, 2001, p. 1506

Rank Order Percent

Epidemiology of Congenital Heart Disease

- More in males, esp. AS, coarctation, HLHS, pulm and tricusp atresia, TGA
- Exceptions: PDA, ASD and Ebstein's more in females
- 25% of infants with significant cardiac disease have extracardiac anomalies, often multiple, and 1/3 of these (cardiac and extracardiac combined) has an established syndrome

Braunwald 7th ed. Ch. 56. 2005;p.1490.

Five T's

Truncus always has increased pulmonary blood flow, the other T's usually have increased blood flow but can be decreased with high PVR, the two E's have decreased pulmonary flow

1. **Tetralogy of Fallot** (most common cyanotic lesion after 1 y.o.)
2. **Tricuspid Atresia**
3. **Transposition of the Great Arteries**
4. **Truncus Arteriosus**
5. **Total Anomalous Pulmonary Venous Return**

Two E's
Ebstein's Anomaly
Eisenmenger Syndrome

Braunwald 7th ed. Ch. 56. 2005;p.1495.

Ductal-Dependent Lesions

- D-TGA
- HLHS

Causes of Cyanotic Heart Disease

Truncus always has increased pulmonary blood flow, the other T's usually have increased blood flow but can be decreased with high PVR, the two E's have decreased pulmonary flow

Perloff, 1994. p. 5 (not included in 2003 ed)

CYANOTIC

Increased Pulmonary Arterial Blood Flow

1. Complete transposition of the great arteries
2. Taussig-Bing anomaly
3. Truncus arteriosus
4. Total anomalous pulmonary venous connection
5. Univentricular heart with low pulmonary vascular resistance and no pulmonary stenosis
6. Common atrium
7. Fallot's tetralogy with pulmonary atresia and increased collateral arterial flow
8. Tricuspid atresia with nonrestrictive ventricular septal defect
9. Complete interruption of the aortic arch with ventricular septal defect and patent ductus arteriosus

Normal or Decreased Pulmonary Arterial Blood Flow

1. Dominant left ventricle
   a. Tricuspid atresia
   b. Pulmonary atresia with intact ventricular septum
   c. Ebstein's anomaly of the tricuspid valve
   d. Single morphologic left ventricle with pulmonary stenosis or high pulmonary vascular resistance
2. Dominant right ventricle
   a. *No pulmonary hypertension*
      i. Pulmonary stenosis or atresia with ventricular septal defect (Fallot's tetralogy)
      ii. Pulmonary stenosis with intact ventricular septum and right to left interatrial shunt
      iii. Pulmonary stenosis with complete transposition of the great arteries
      iv. Double outlet right ventricle with pulmonary stenosis
   b. *Pulmonary hypertension*
      i. Atrial septal defect with reversed shunt
      ii. Ventricular septal defect with reversed shunt
      iii. Patent ductus arteriosus or aortopulmonary window with reversed shunt
      iv. Double outlet right ventricle with high pulmonary vascular resistance
      v. Complete transposition of the great arteries with high pulmonary vascular resistance
      vi. Total anomalous pulmonary venous connection with high pulmonary vascular resistance
      vii. Hypoplastic left heart (aortic atresia, mitral atresia)
3. Normal or nearly normal ventricles
   a. Pulmonary arteriovenous fistula
   b. Vena caval to left atrial communication

Cyanosis

- Excess concentration of circulating reduced hemoglobin, over 3-4 gm/dl*
- Central cyanosis is characteristic of shunt, peripheral cyanosis (“acrocyanosis”) indicates excessive extraction and peripheral constriction, a patient can show both*
- Hypoxemia stimulates renal oxygen sensors to increase erythropoetin, so hemoglobin concentration is increased
- Increased hemoglobin concentration compensates for low arterial oxygen saturation (increased oxygen delivery), thus it is adaptive

*Braunwald, 2001, p. 1513, 1617

Hyperviscosity Syndrome

- Symptoms: mainly CNS and usually stereotypic for an individual patient - headache, altered mentation, visual disturbances, tinnitus, paresthesias, fatigue, dizziness, and myalgias; relief by phlebotomy is defining
- Usually hematocrit is >65% with symptoms, may be less if iron deficient or dehydration (excessive heat, illness, fever, diarrhea, vomiting)
- Asymptomatic elevation in hematocrit is not an indication for phlebotomy (unless preoperative and hct >65 to decrease risk of perioperative hemorrhage, then could save for autologous transfusion)
- Phlebotomy: remove 250-500 ml over 30-45 minutes preceded by or simultaneous with quantitative NS (Dextran if CHF) replacement; iron supplementation; repeat QD till symptomatic improvement or Hb has “fallen too far”

Braunwald, 2005, p. 1496

Iron Deficiency

- Common and important in cyanosis, from hemoptysis, epistaxis, menses, inappropriate phlebotomy
- Microcytosis increases whole blood viscosity (less deformable than biconcave disc)
- Replace iron till hematocrit increases or till iron-replete state, IV iron for oral intolerance

Braunwald, 2001, p. 1617

Abnormality in Hemostasis

- Elevated PT and PTT, decreased levels of factors V, VII, VIII, and IX, qualitative and quantitative platelet disorders, increased fibrinolysis*
- Spontaneous superficial bleeding is usually self-limited; avoid ASA, NSAID and heparin
- Hemoptysis or intracranial or GI bleeding are concerns
- Anticoagulant usually should be avoided, but in atrial fibrillation or mechanical prosthesis, a risk-benefit dilemma must be addressed

Braunwald, 2001, p. 1617; *J Cardiothorac Vasc Anesth 2002;16:752

Cerebrovascular Events

- Stroke from cerebral arterial thrombosis usually seen in patients with iron deficiency or iron depletion
- Cerebral hemorrhage with anticoagulant therapy
- Paradoxical emboli occur in R>L shunt, either thrombus or air from IV line without a filter
- Brain abscess may present with headache and fever and focal finding or seizure

Braunwald, 2005, p. 1496

Arthralgia

- Hypertrophic osteoarthropathy is usual cause – arthralgias and bone pain
  - Affects up to 1/3 patients with cyanotic congenital heart disease
  - Mechanism: megakaryocytes from marrow bypass lung and lodge in arterioles and capillaries and induce release of PDGF promoting local cell proliferation – new osseous formation with periostitis
- Gouty arthritis – treatable with colchicine, probenecid, antiinflammatory agents or allopurinol

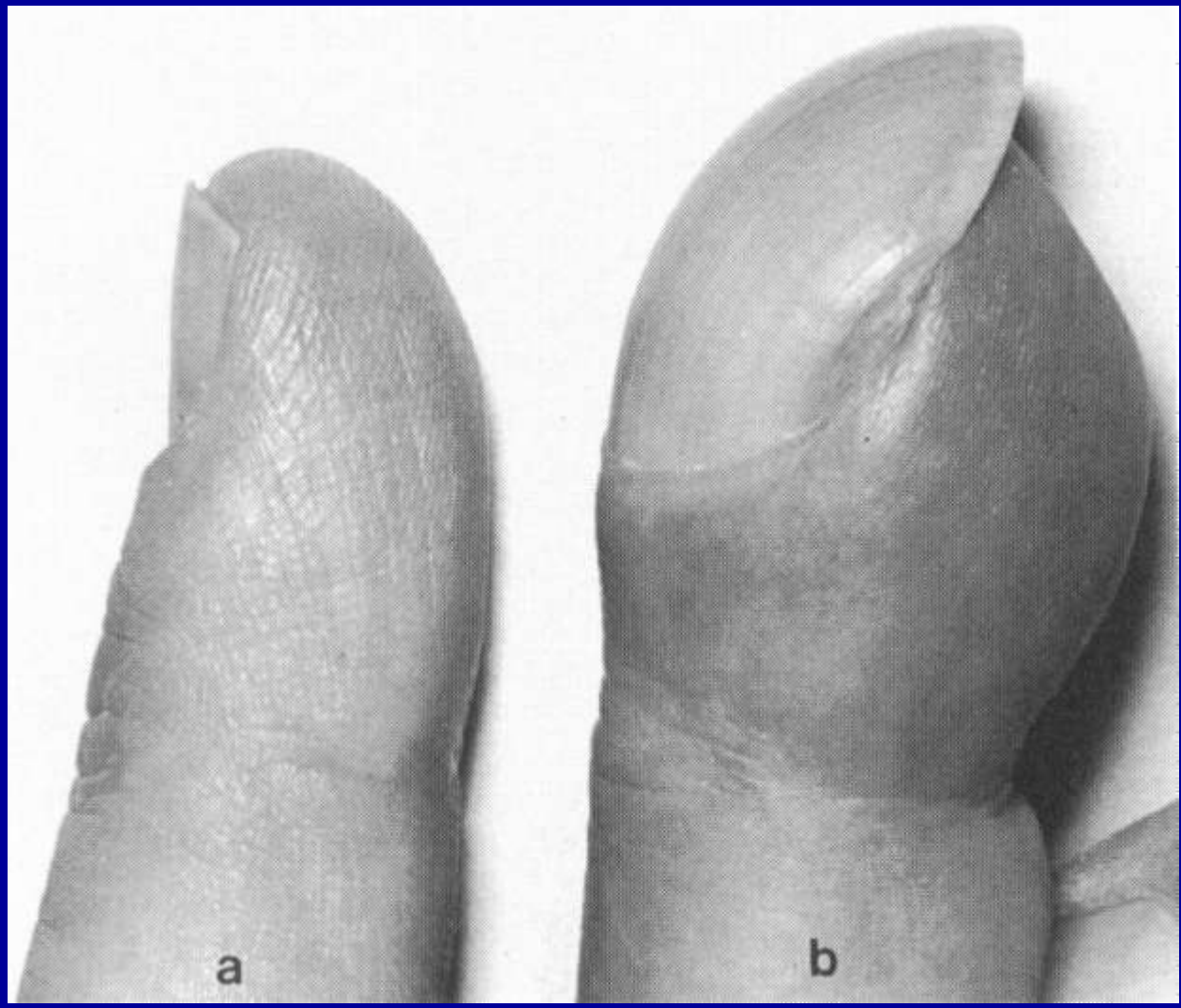
Braunwald, 2005, p. 1496

Clubbing

- *Characteristic of central cyanosis (cardiac or pulmonary disease with hypoxia, also can appear in infective endocarditis)
- *Early – increased glossiness and cyanosis of skin at nail root
- *Obliteration of the normal angle between nail base and skin, then hypertrophy of the pulp soft tissue, nail root floats freely, palpable loose proximal end of nail
- Increased number of capillaries, increased blood flow, extensive AV aneurysms and increase in connective tissue
- PDA and Eisenmenger physiology, clubbed toes, differential cyanosis
- PDA and TGA and Eisenmenger physiology, clubbed fingers, differential cyanosis

*Braunwald, 2005, p. 78-9. Braunwald, 2001.

Normal versus Clubbing



Perloff, 1994, p. 7

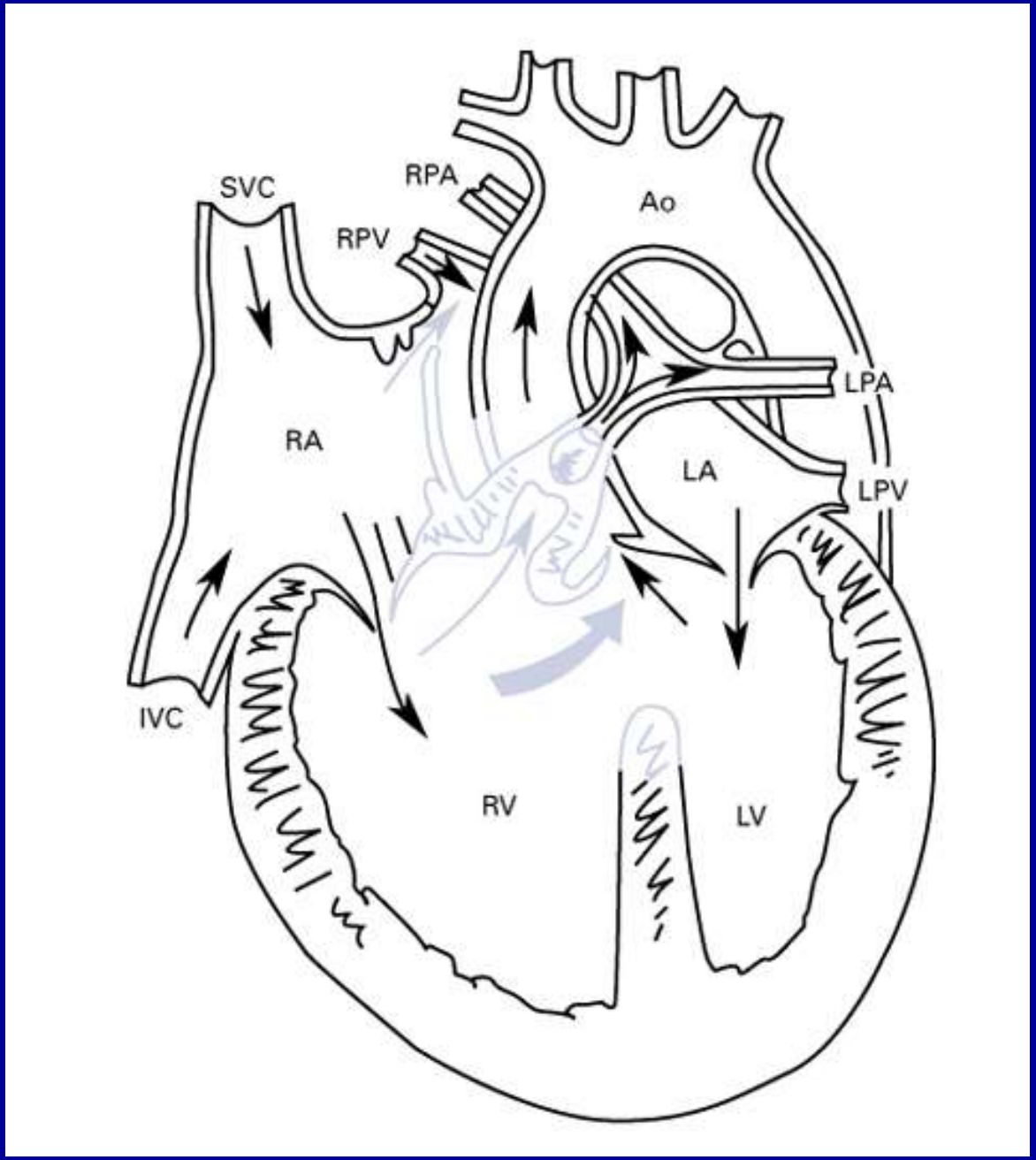
1. Tetralogy of Fallot

Tetralogy of Fallot

- 1888 la maladie bleue – Etienne-Louis Arthur Fallot, diagnosed at bedside
- VSD + RVOTO + overriding aorta + RVH
- Cause: anterior deviation of septal insertion of the infundibular ventricular septum

Classic Tetralogy of Fallot

- RVH
- Overriding aorta
- RVOTO, infundibular, PV also usually involved
- VSD, usually perimembranous due to fibrous continuity with TV and AoV, lies subarterial

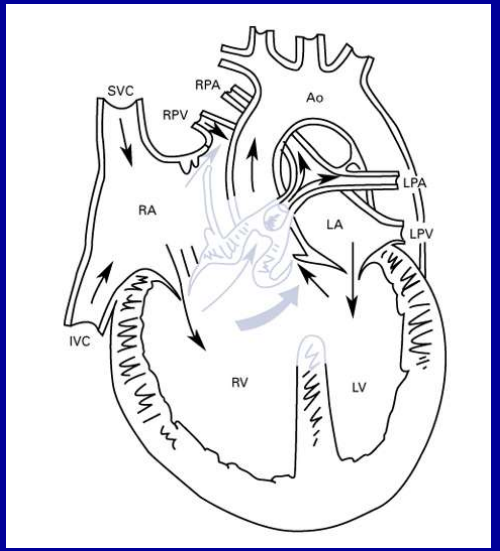


From Hurst, 1999, Ch 70

Tetralogy of Fallot, Associated Lesions

- Coronary artery anomaly, LAD from RCA and anterior course in 5%
- Right-sided Aortic arch in up to 25%, more if more cyanotic
- ASD in maybe 15% of patients (ASD=pentalogy of Fallot)

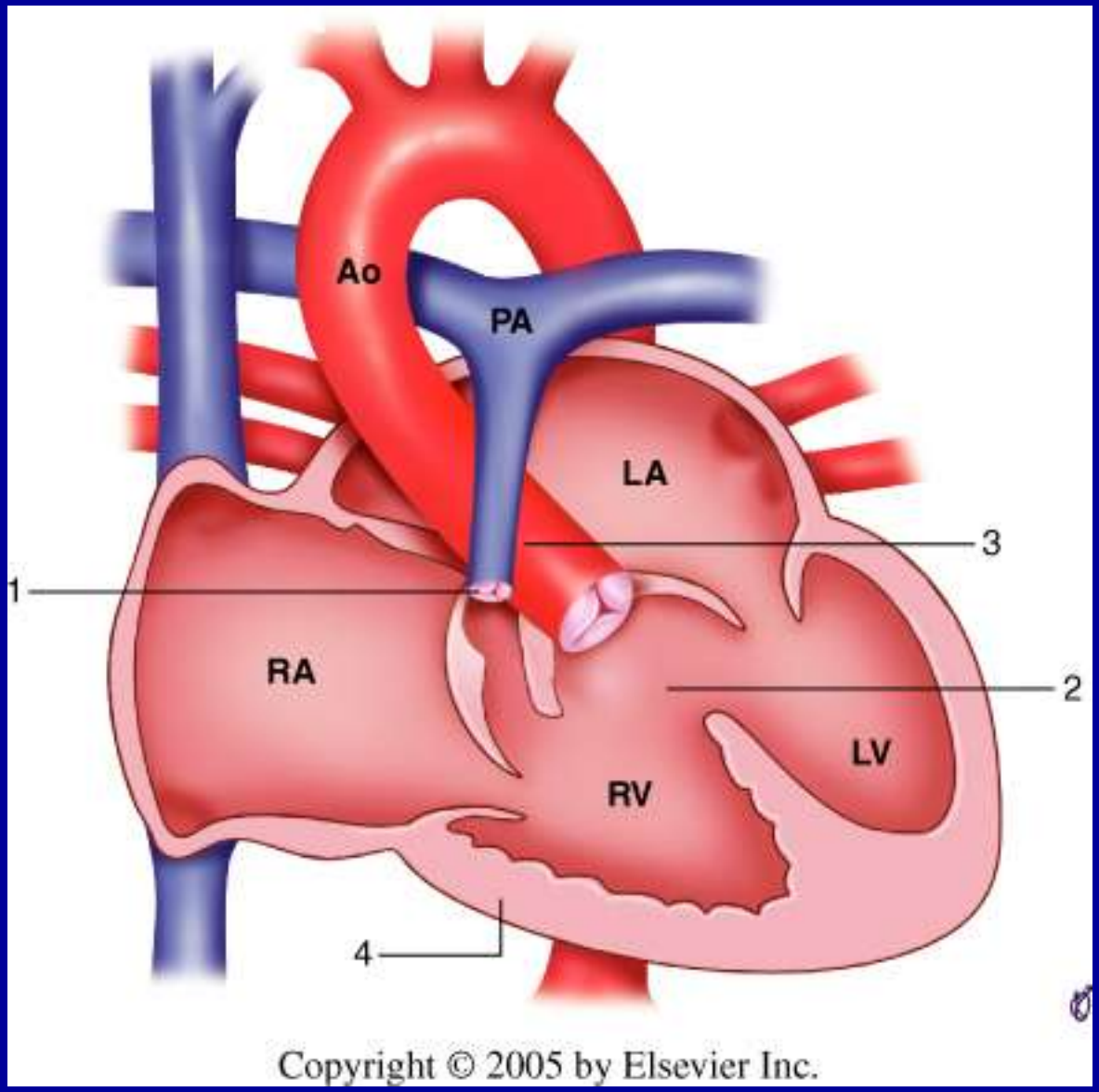
Tetralogy of Fallot



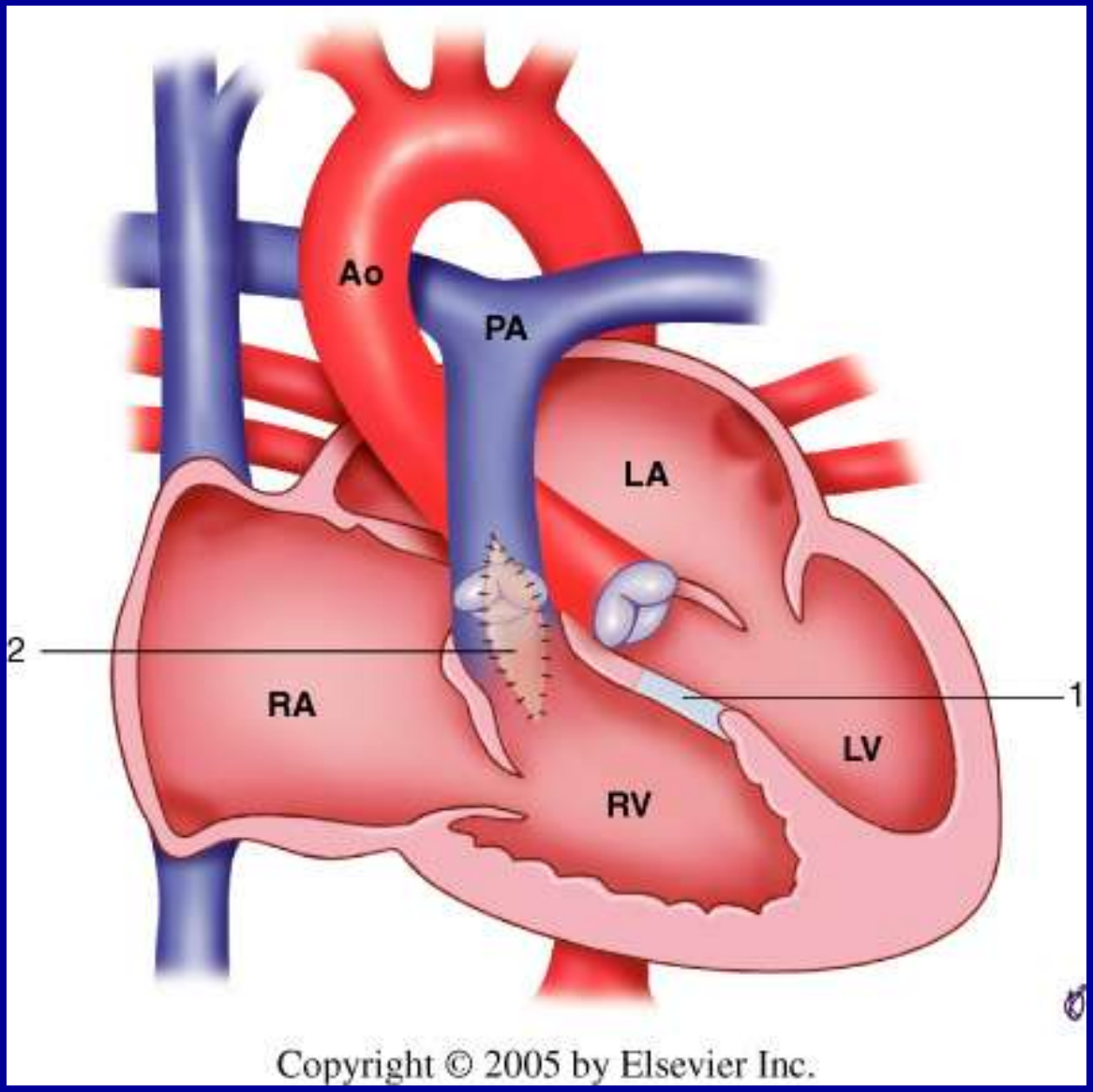
- Severity of manifestations generally related to extent of RVOTO
- Generally RVSP=LVSP
- Murmur is RVOTO, VSD is silent, S2 is single
- "Pink tet" has less RVOTO, spectrum of RVOTO extends to pulmonic valve atresia
- Exercise deepens cyanosis from decreased systemic vascular resistance – squat, spells (murmur softens during spell due to less PBF)
- Exam after complete repair: pulm regurg, single S2, residual PS murmur, possible residual VSD

Moss and Adams, p. 888

Tetralogy of Fallot Anatomy

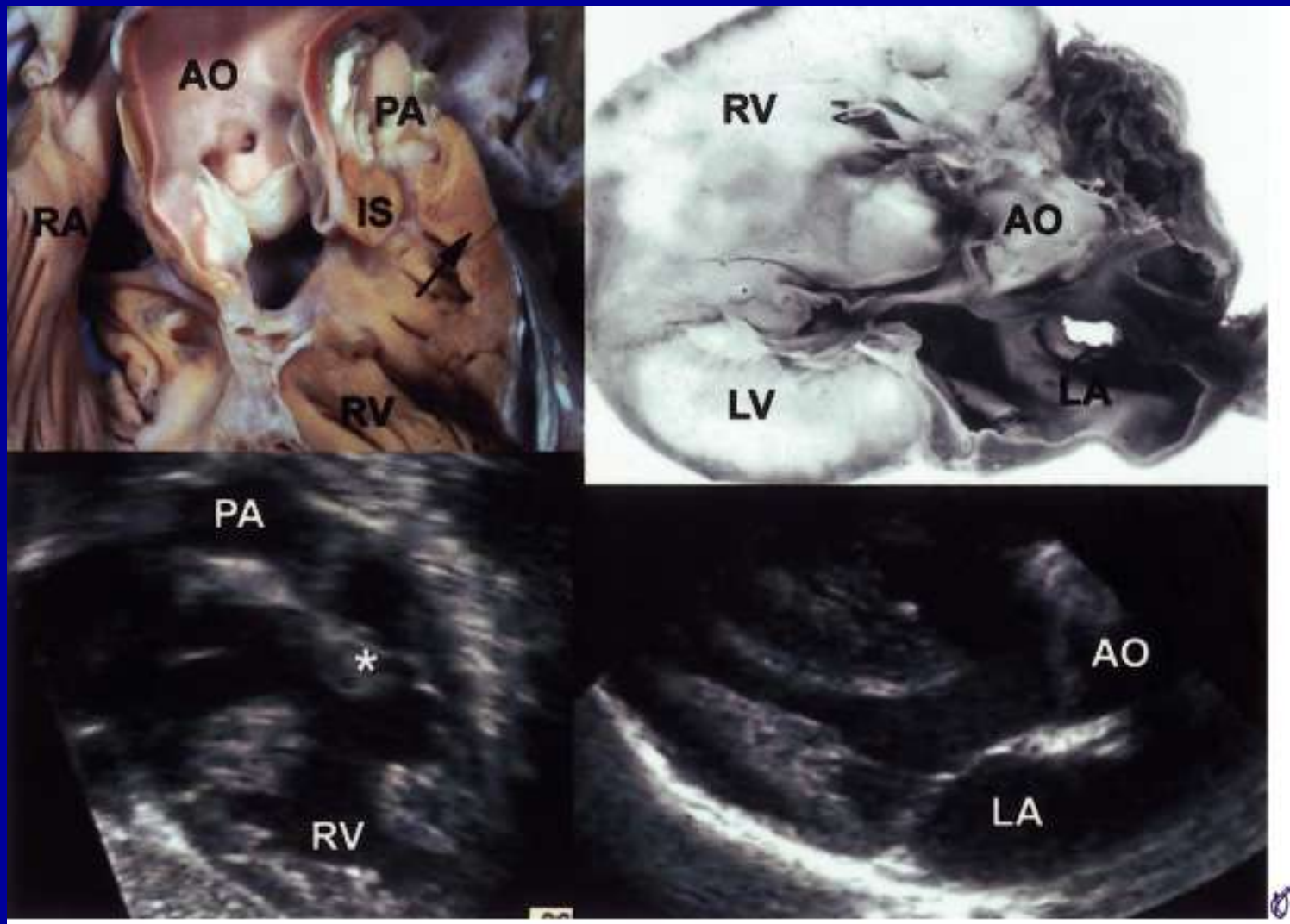


Tetralogy of Fallot – Surgical Repair



Tetralogy of Fallot

Hypertrophied septoparietal trabeculations



Surgery for Tetralogy of Fallot

- Initial palliation now infrequently done, was to increase pulmonary blood flow (SA to PA)
  - Blalock-Taussig (first done 1945 on a patient with TOF); modified with Gore-Tex
  - May use with severe pulmonary artery hypoplasia (lack of flow results in lack of development) or aberrant coronary
  - Waterston and Potts are largely of historical interest – they had pulmonary artery distortion and inconsistent results of flow and pressure effects
- Complete repair: Relieve RVOTO, try not to destroy PV, maybe atrial approach; close VSD, close ASD if present
  - Significant aortopulmonary collateral artery flow or PDA may affect surgical decisions

Moss and Adams, p. 895

Aortopulmonary Shunts (SA to PA)

TABLE 9-1

Aortopulmonary Shunts

Shunt	Surgeon	Year
Blalock-Taussig shunt	Alfred Blalock	1944
Potts shunt	Willis Potts	1946
Waterston shunt	David Waterston	1962
Cooley shunt	Denton Cooley	1966
Modified Blalock-Taussig shunt	Marc de Leval	1976

*

*Cooley is similar to Waterston but intrapericardial anterior approach

Mavroudis, 2003, p. 161

TABLE 56–5 Palliative Systemic-to-Pulmonary Shunts

Arterial

Blalock-Taussig shunt (subclavian artery to PA)
- Classic—end-to-side, no or reduced ipsilateral arm pulses
- Current—side-to-side tubular grafts, preserved arm pulses

Central shunt (side-to-side tubular graft, aorta to PA)

Potts shunt (descending aorta to LPA)

Waterston shunt (ascending aorta to RPA)

Venous

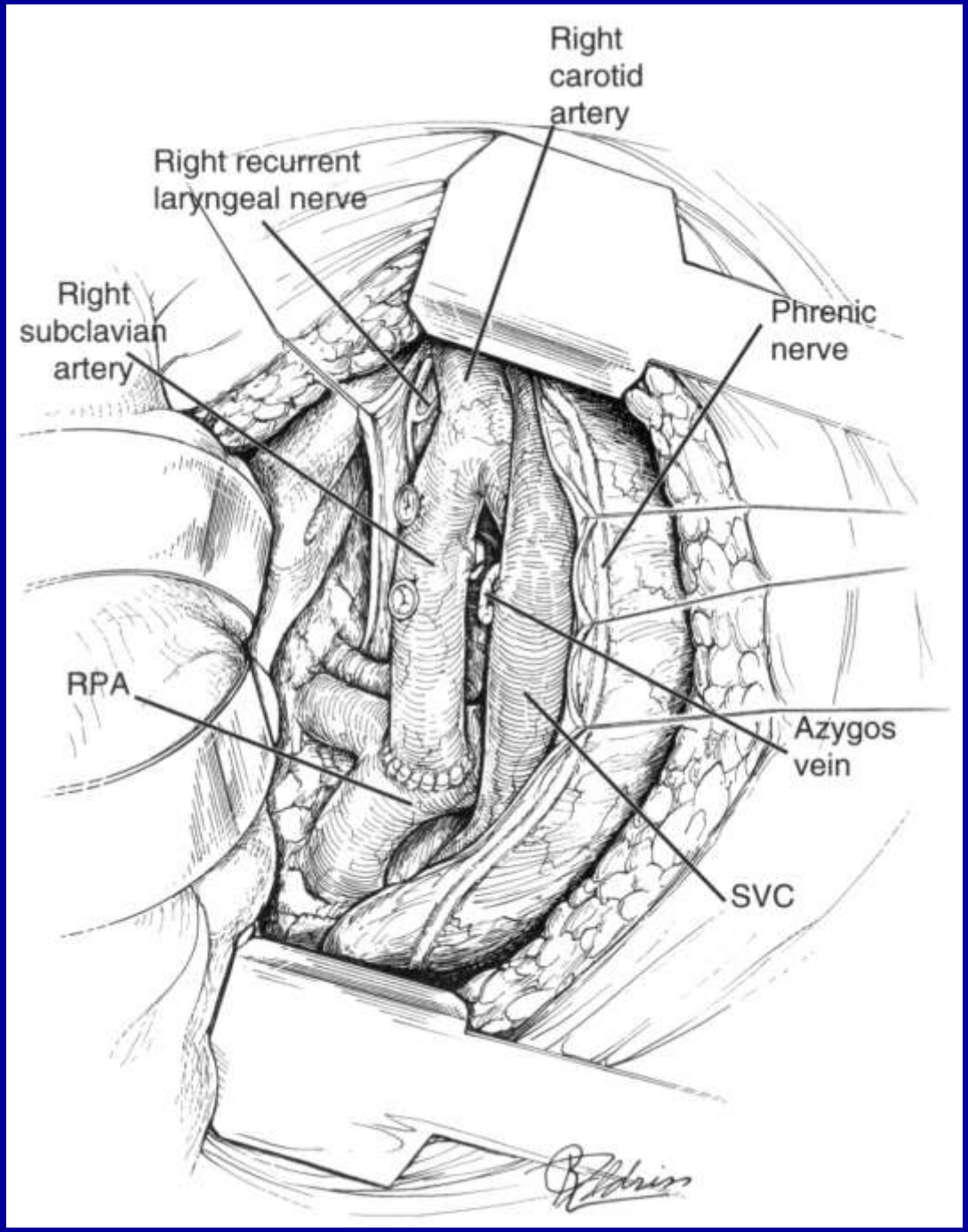
Glenn shunt (SVC to ipsilateral PA without cardiac or other PA connection)

Bidirectional cavopulmonary (Glenn) shunt (end-to-side SVC to LPA and RPA shunt)

PA = pulmonary artery; LPA = left PA; RPA = right PA; SVC = superior vena cava.

Classic Blalock-Taussig

Tie off the right subclavian distally, so right arm has decreased blood flow and can have some long term sequelae



Mavroudis, 2003, p. 164

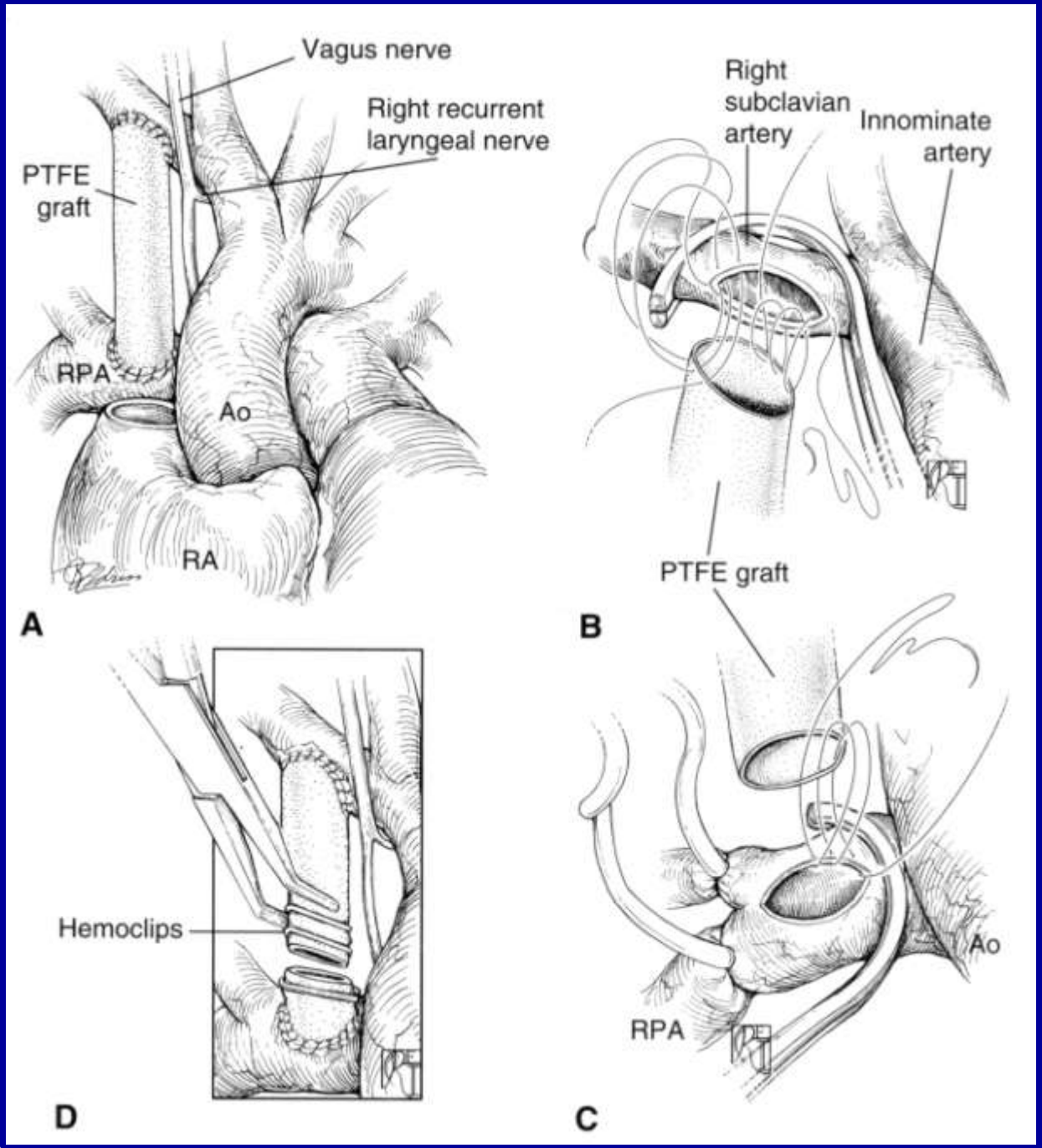
Modified Blalock-Taussig

Issue: selection of right size of graft diameter

A – anatomy of completed repair

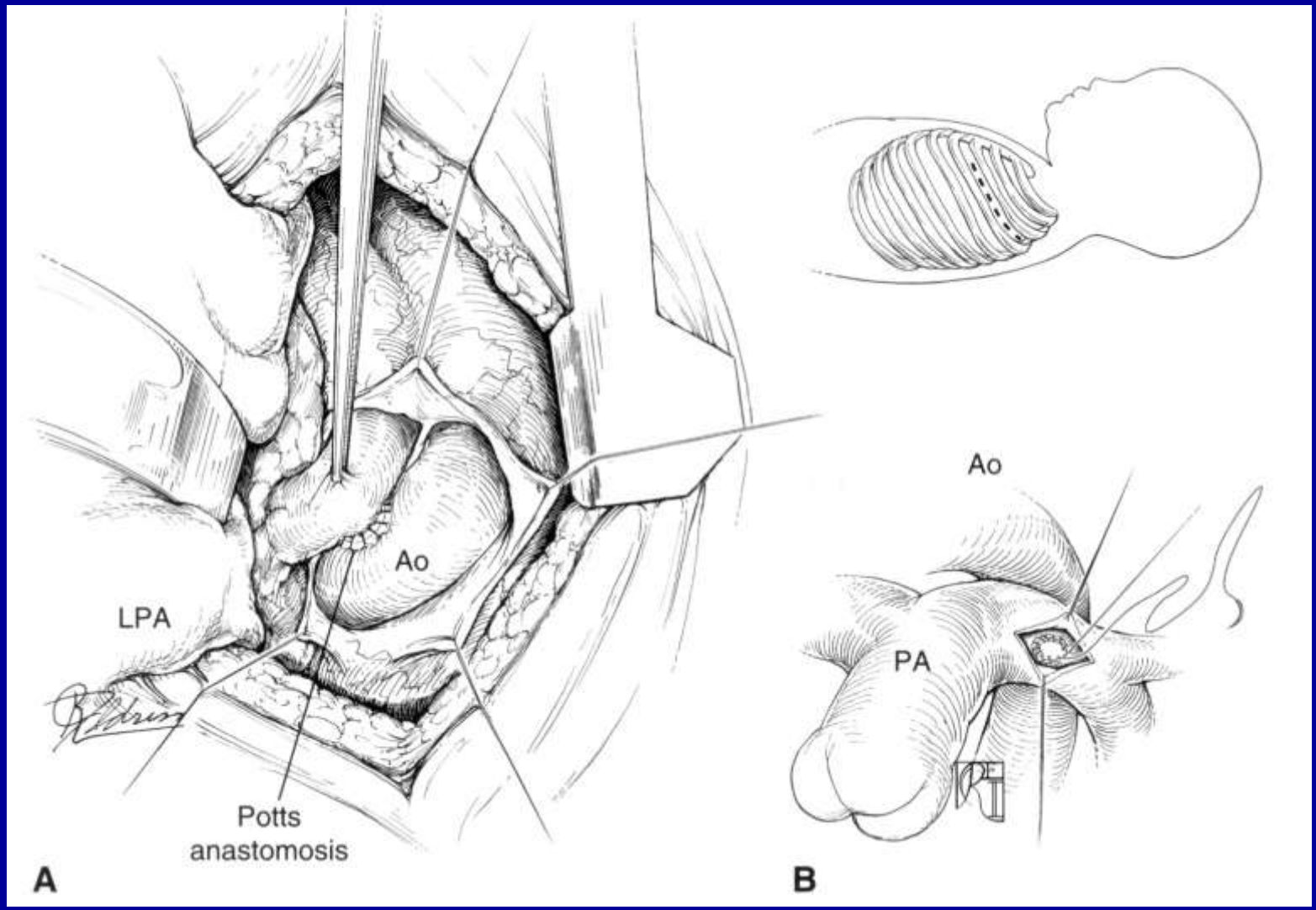
B and C – technique of repair

D – taking down the repair



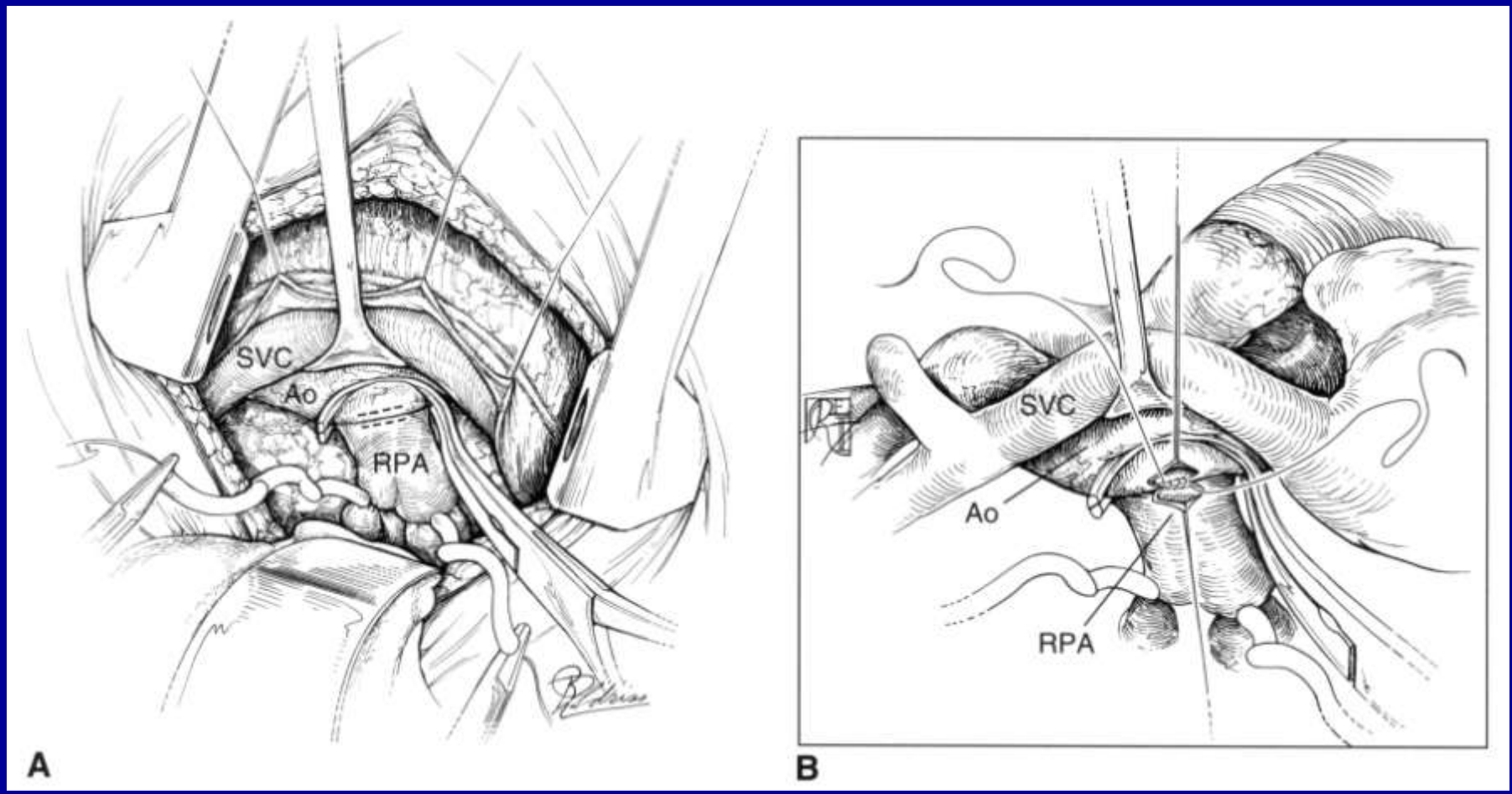
Mavroudis, 2003, p. 165

Potts Shunt



Mavroudis, 2003, p. 169

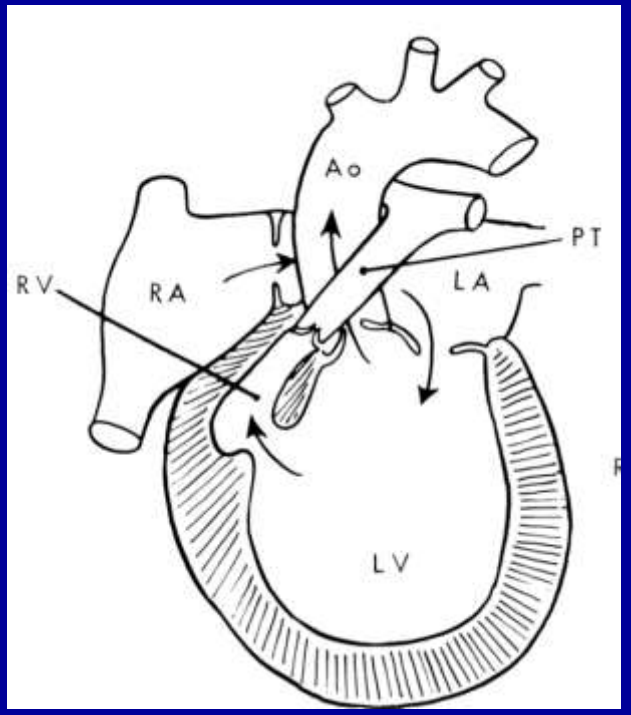
Waterston Shunt



Mavroudis, 2003, p. 167

2. Tricuspid Atresia

Tricuspid Atresia



- TV is represented by a dimple in the RA floor, muscular or fibrous membrane
- Obligate interatrial communication: PFO (usually restrictive) or secundum ASD, rarely primum ASD
- Obligate systemic to pulmonary communication, usually membranous VSD (if pulmonary atresia, PDA functions)
- Invariable secondary RV problems: RV inlet is absent, RV trabecular portion is incomplete, and infundibular portion remains – size of VSD is related to size of RV, and size of VSD also related to size of PV

Moss and Adams, 2001, p. 799

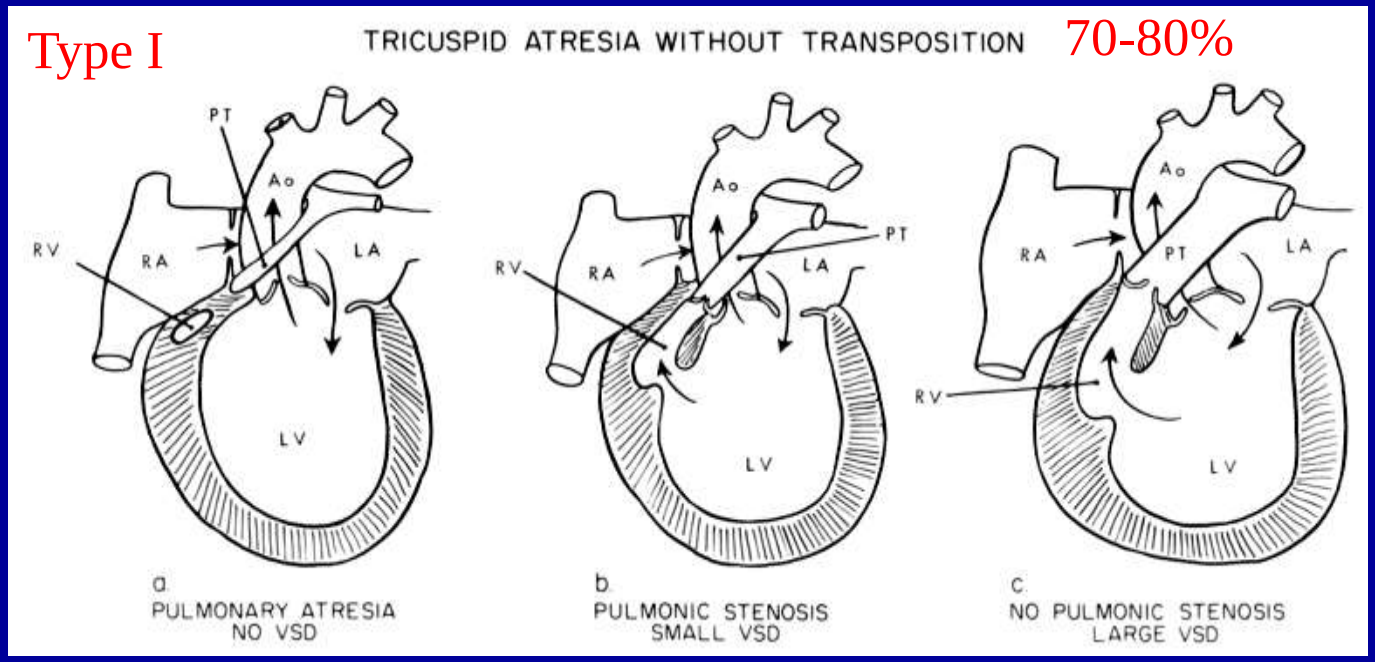
Tricuspid Atresia

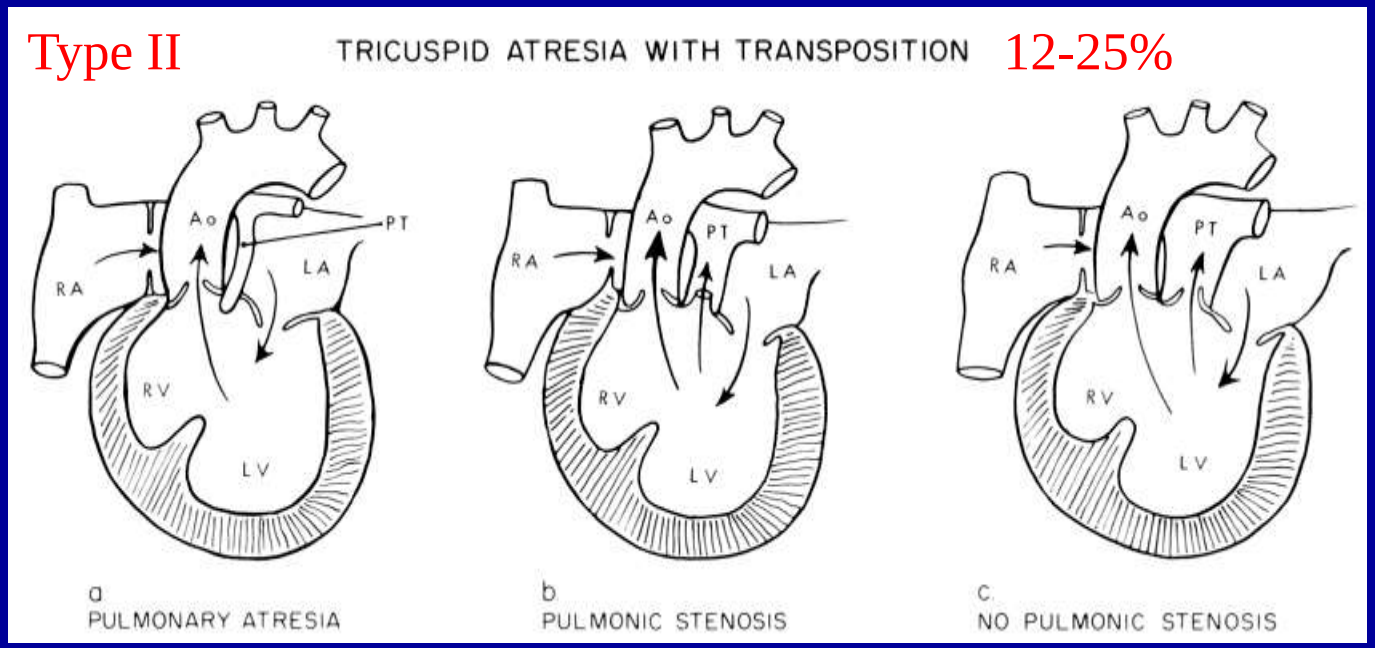
All have complete admixture of venous returns

Cyanosis at least by 1 week

Functional single ventricle

Type III, uncommon, used for more complex great artery problems





Perloff, 1994, p. 616, Moss and Adams, 2001, p. 800

Types of Tricuspid Atresia

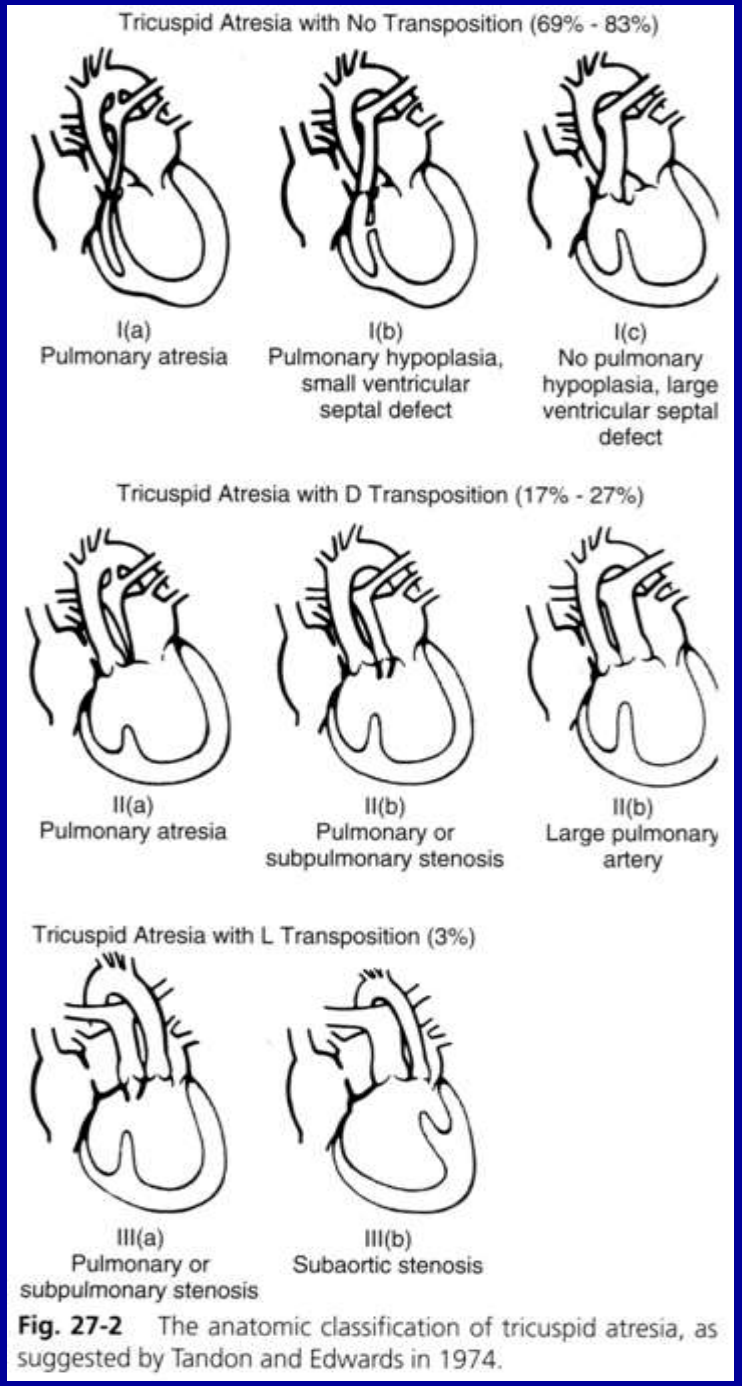
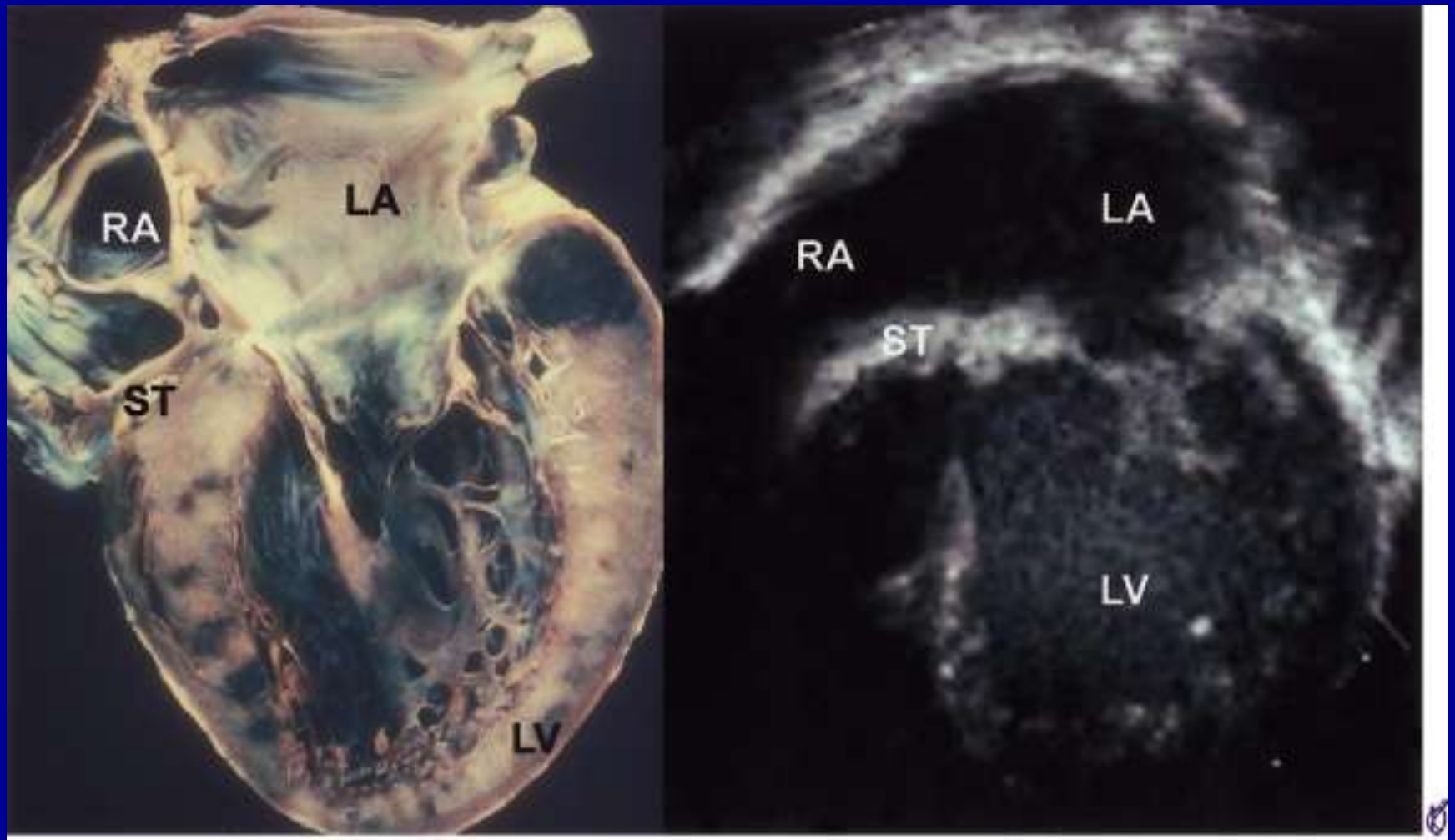


Fig. 27-2 The anatomic classification of tricuspid atresia, as suggested by Tandon and Edwards in 1974.

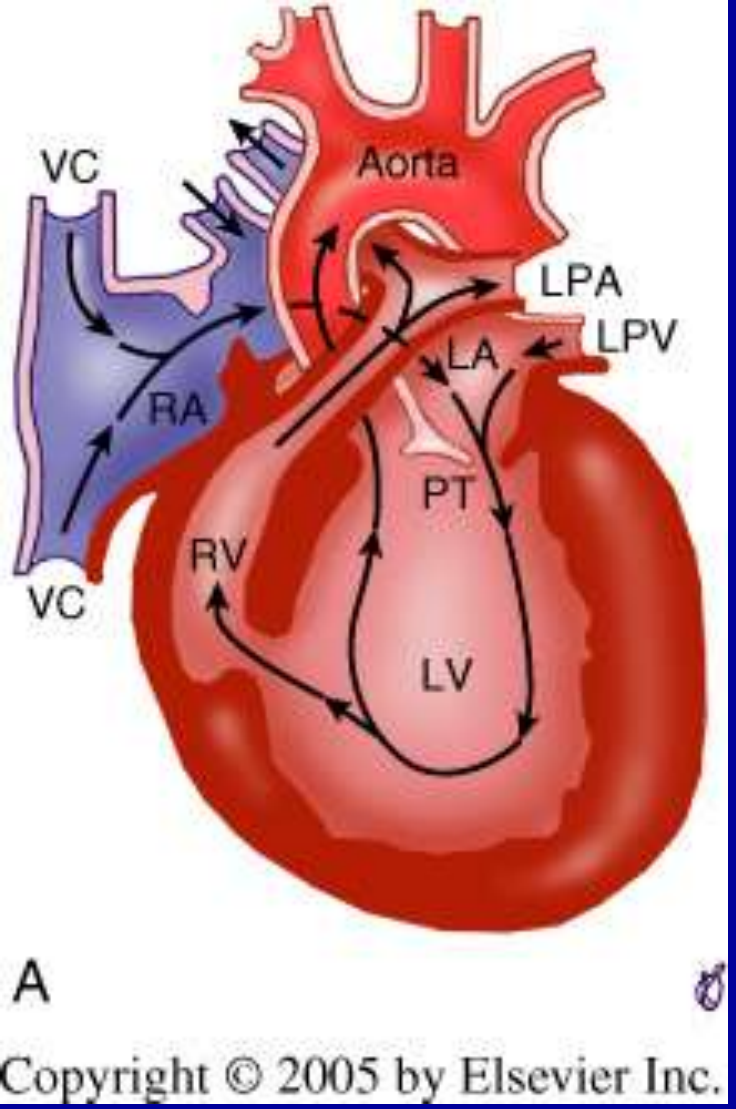
Mavroudis 2003, p. 499

Tricuspid Atresia (univentricular connection of the LV type with absent right connection)

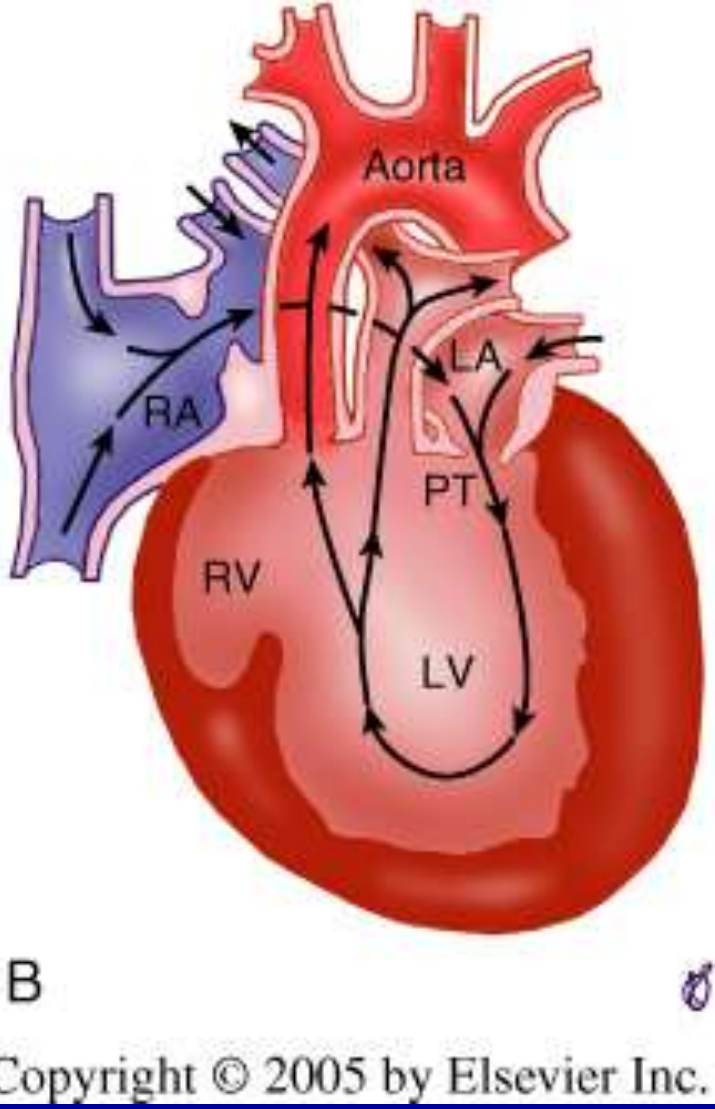


Wedge of (ST) sulcus tissue in the floor of the RA

Tricuspid Atresia



No TGA, small VSD, small RV, narrow RVOT



TGA, large VSD (essentially a common ventricle), aorta arising from an infundibular component of the RV

Tricuspid Atresia Treatment

- Create connection from systemic vein to PA and eliminate atrial shunt
- At birth, diminished pulmonary blood flow is an indication for Pg E1 to keep ductus patent till surgery of aortopulmonary shunt
- If associated transposition and no pulm flow obstruction, may need pulmonary banding

Adams and Moss, 2001, p. 807

Systemic Vein to PA Anastomosis

- Glenn, 1965, SVC to distal RPA (residual R-L shunt from IVC)
- Bidirectional Glenn – SVC end-to-side to RPA, maintains PA continuity, largely has replaced original
- Fontan and Baudet, 1971, SVC to RPA and RA appendage to LPA and aortic homograft, and close ASD
- Kreutzer, RA to MPA with interposition of semilunar valve (PA branches maintain mutual continuity)
- Fontan operation now can refer to any operation that connects systemic vein and pulmonary artery without ventricular passage, may be done after a Glenn

Adams and Moss, 2001, p. 807, Mavroudis, 2003, p. 504

Criteria Indicating Fontan Success

- Age 4-15 years (younger now)
- NSR
- Normal systemic venous connections (less now)
- Normal RA size
- Normal PA pressure (mean <15mmHg)
- Low PVR (<4 Woods units/m2)
- Adequate PA diameter (>75% Ao)
- **LVEF >60%
- **No MR
- No complicating factors (prior surgery, PA distortion)

Adams and Moss, 2001, p. 807

** current relative contraindications

Fontan Original Repair

Top: no TGA, insert valve in IVC, band the PA

Bottom: with TGA, insert valve in IVC and RPA, band the PA

Today, valves are avoided because they cause more problems

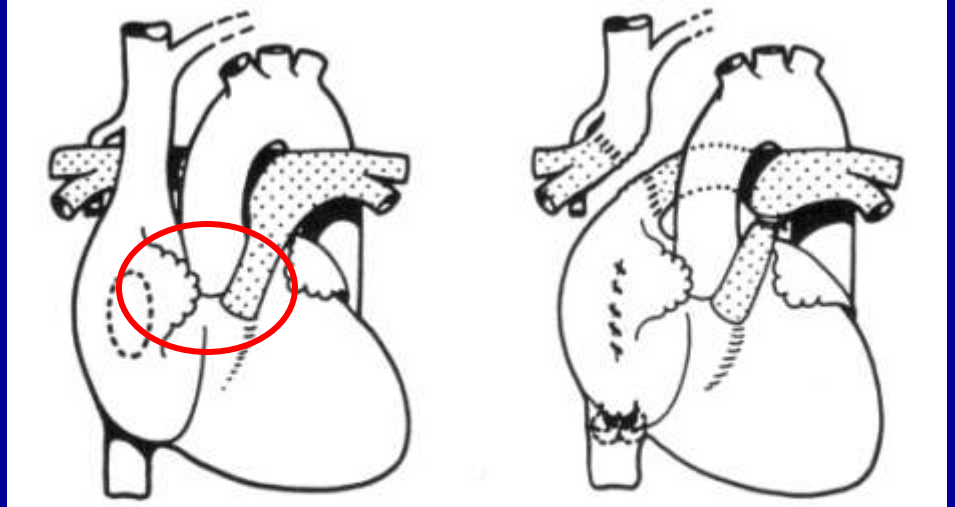


Fig. 27-5 Artist's drawing of Fontan's original repair for tricuspid atresia type Ib. Case 1.

(From Fontan F, Baudet E: *Thorax* 26:240, 1971.)

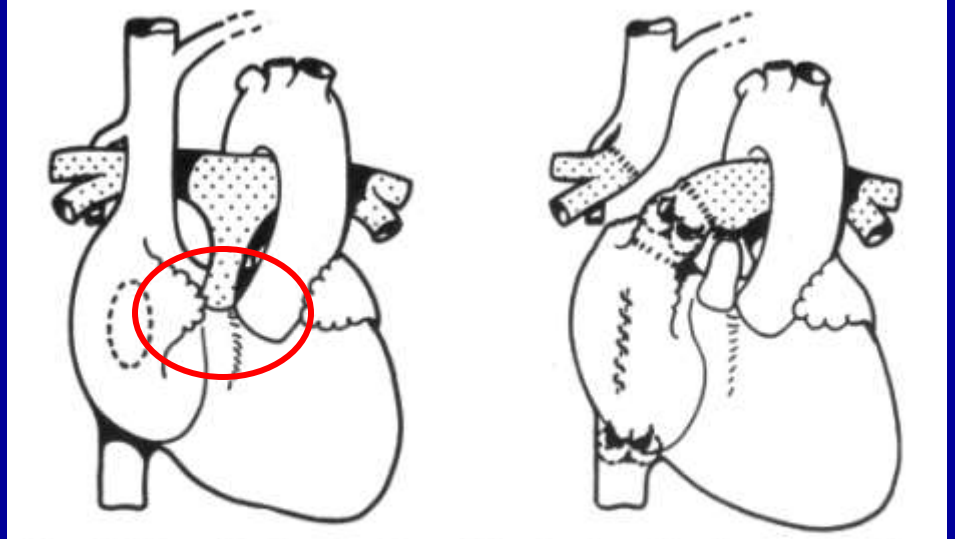
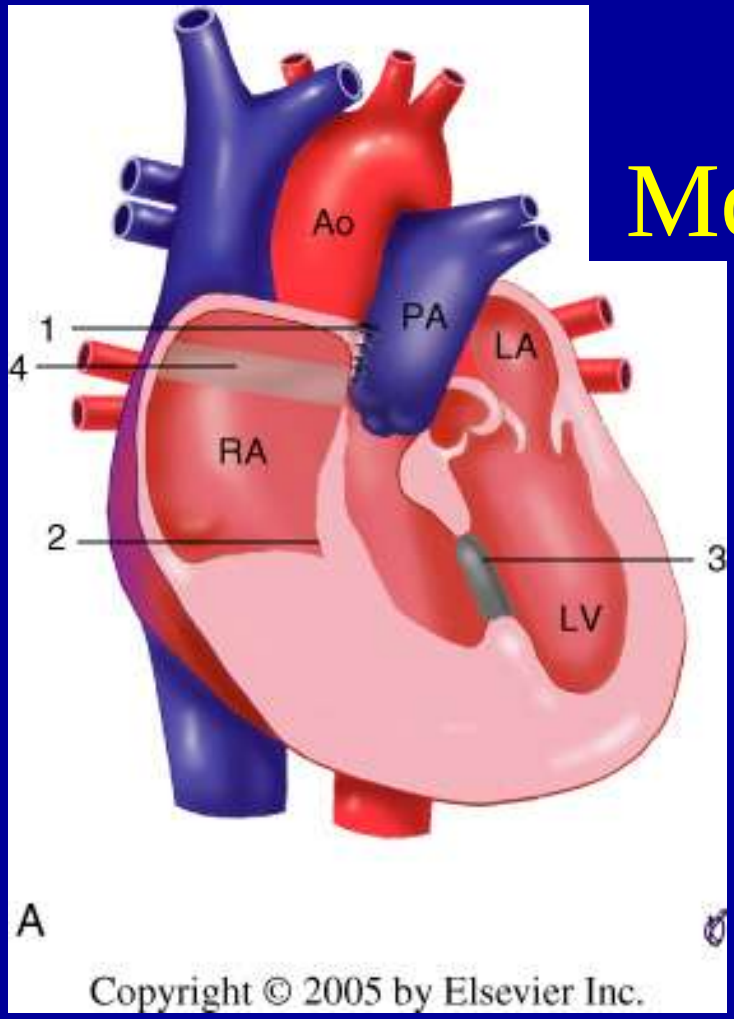


Fig. 27-6 Artist's drawing of Fontan's original operation for repair of tricuspid atresia type IIb.

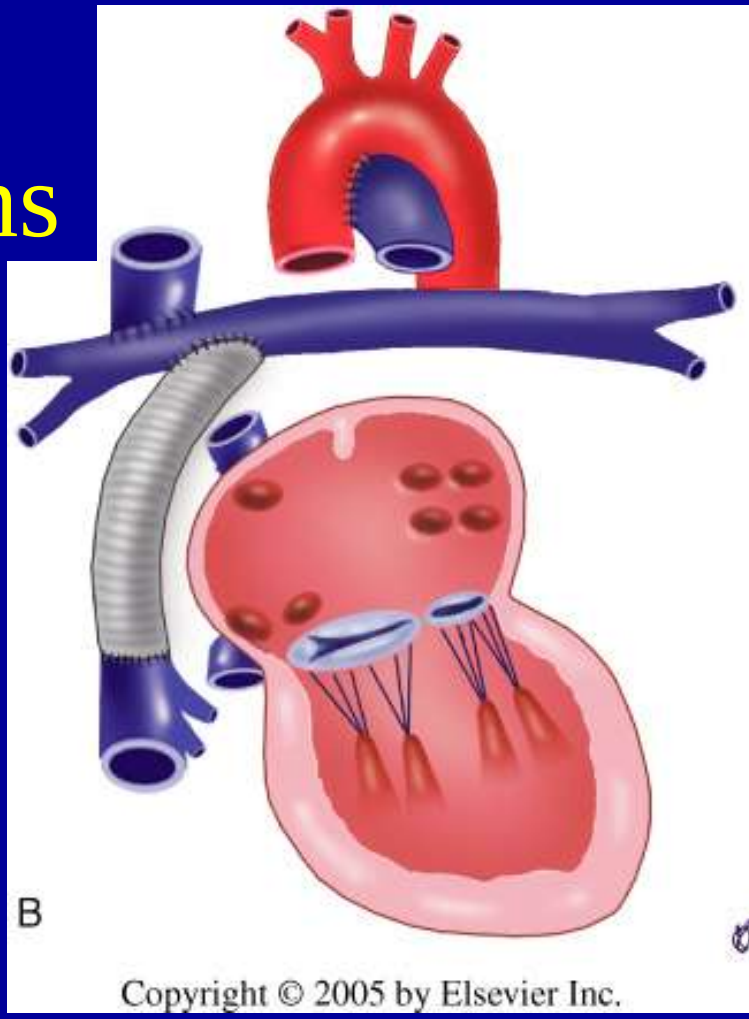
(From Fontan F, Baudet E: *Thorax* 26:240, 1971.)

Mavroudis, 2003, p. 504

Fontan Modifications



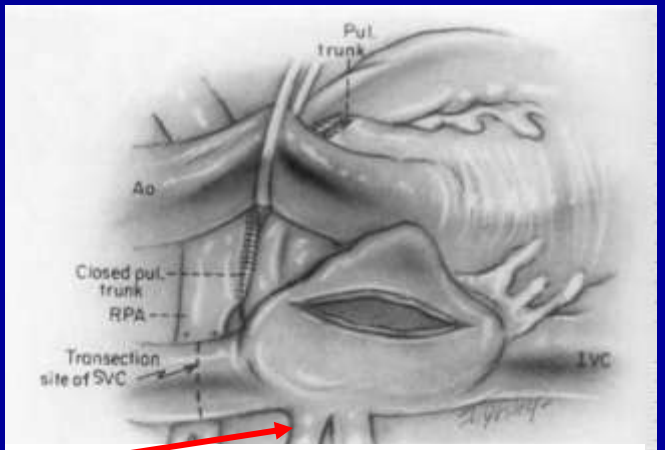
Direct atriopulmonary connection (1) for tricuspid valve atresia (2); ventricular septal defect, oversewn (3); patch closure of atrial septal defect (4).



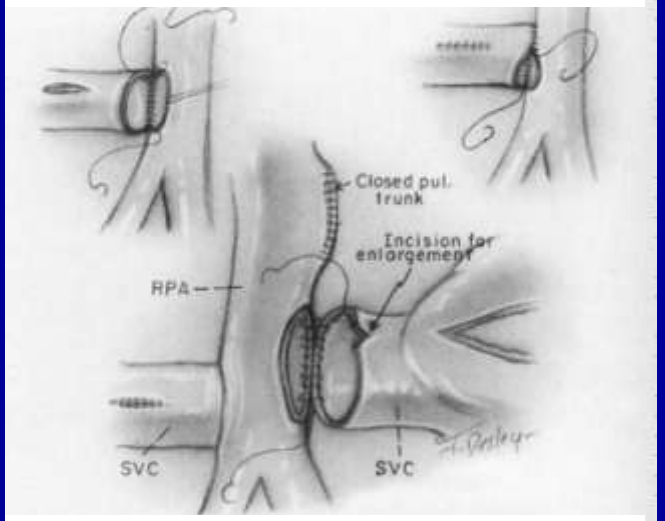
Extracardiac conduit made of a Dacron graft bypassing the right atrium, connecting the inferior vena cava to the inferior aspect of the right pulmonary artery. Superior vena cava is anastomosed to the superior aspect of the right pulmonary artery.

Fontan Operation

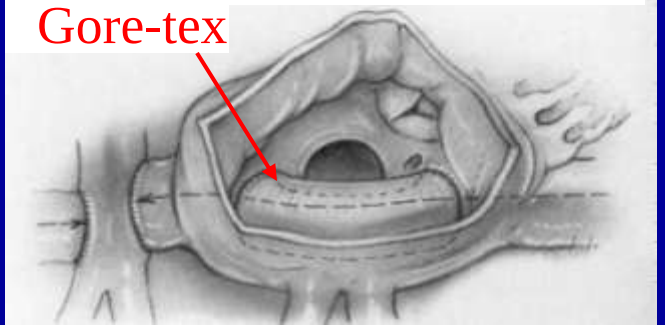
Ant
Sup Inf
Post

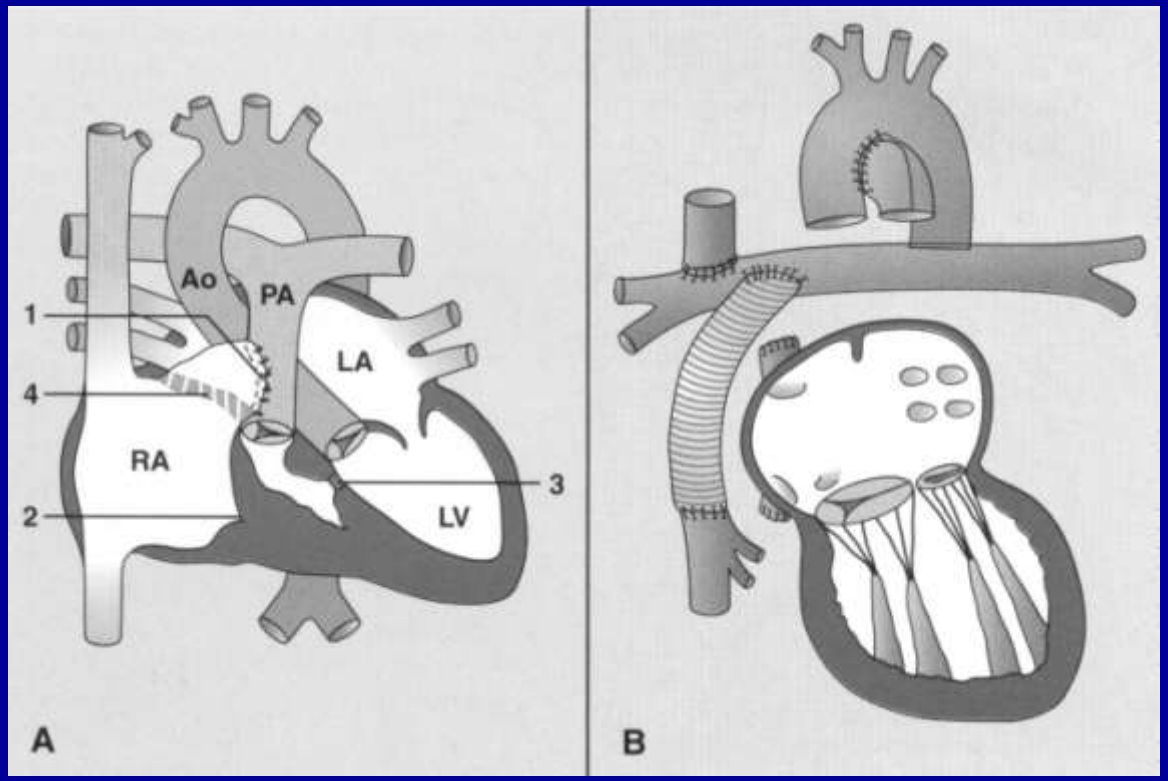


Anomalous pulm vein



A tunnel (gusset) from Gore-tex





Braunwald, 2001, p. 1564, 1607

Results of Fontan

- Unoperated tricuspid atresia has a 1-year mortality of 90%
- Surgical mortality is about 10%
- RA pressure = PA pressure
  - Pleural effusions
  - Low left heart filling if PVR is elevated
  - Protein-losing enteropathy is often the major morbidity
- Exercise capacity remains diminished, may be surprisingly good, considering single ventricle
- Transplantation is an option

Adams and Moss, 2001, p. 808

3. Transposition of the Great Arteries (d-TGA)

Transposition of the Great Arteries (d-TGA)

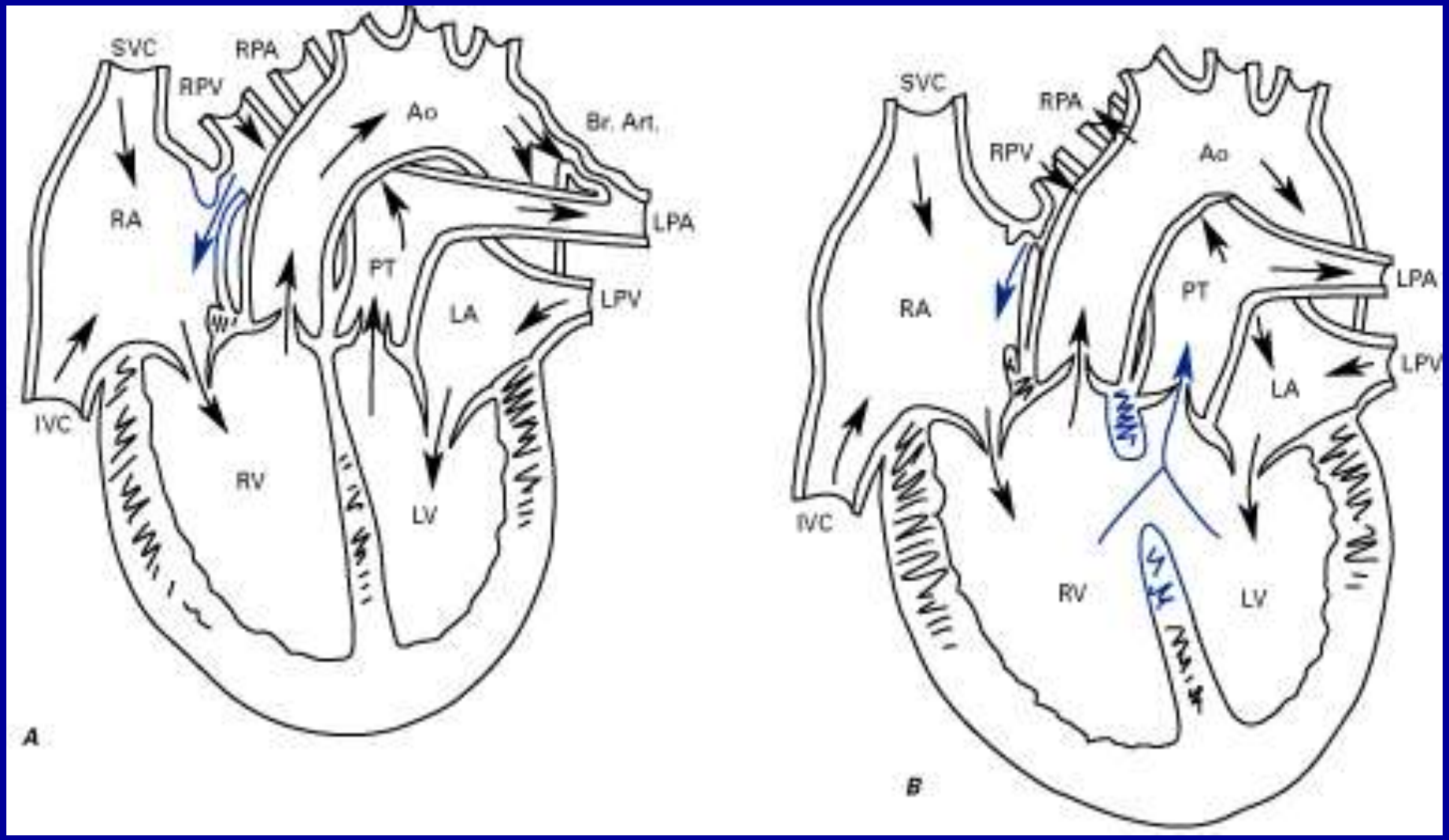
- Lethal and relatively frequent
- The conus (infundibulum) is usually subaortic, right-sided and anterior, preventing fibrous continuity between TV and AoV (but continuity between MV and PV)
- Extensive coronary variability, arise from “facing” coronary sinuses (67% usual, 16% LCX from RCA)

D-TGA Associations

- Nearly half the hearts have no associated anomaly except PFO or PDA
- Most frequent, VSD in 40-45%, small, large, or multiple, 33% membranous, 37% muscular, 30% malalignment (outlet)
- Malalignment VSD associates with overriding of PV onto RV, and if large begin to be DORV, with subpulmonic VSD (Taussig-Bing anomaly), may be associated with LVOTO
- LVOTO in about 25%, more if VSD present, may be dynamic from bulge of IVS into LVOT (subpulmonic)

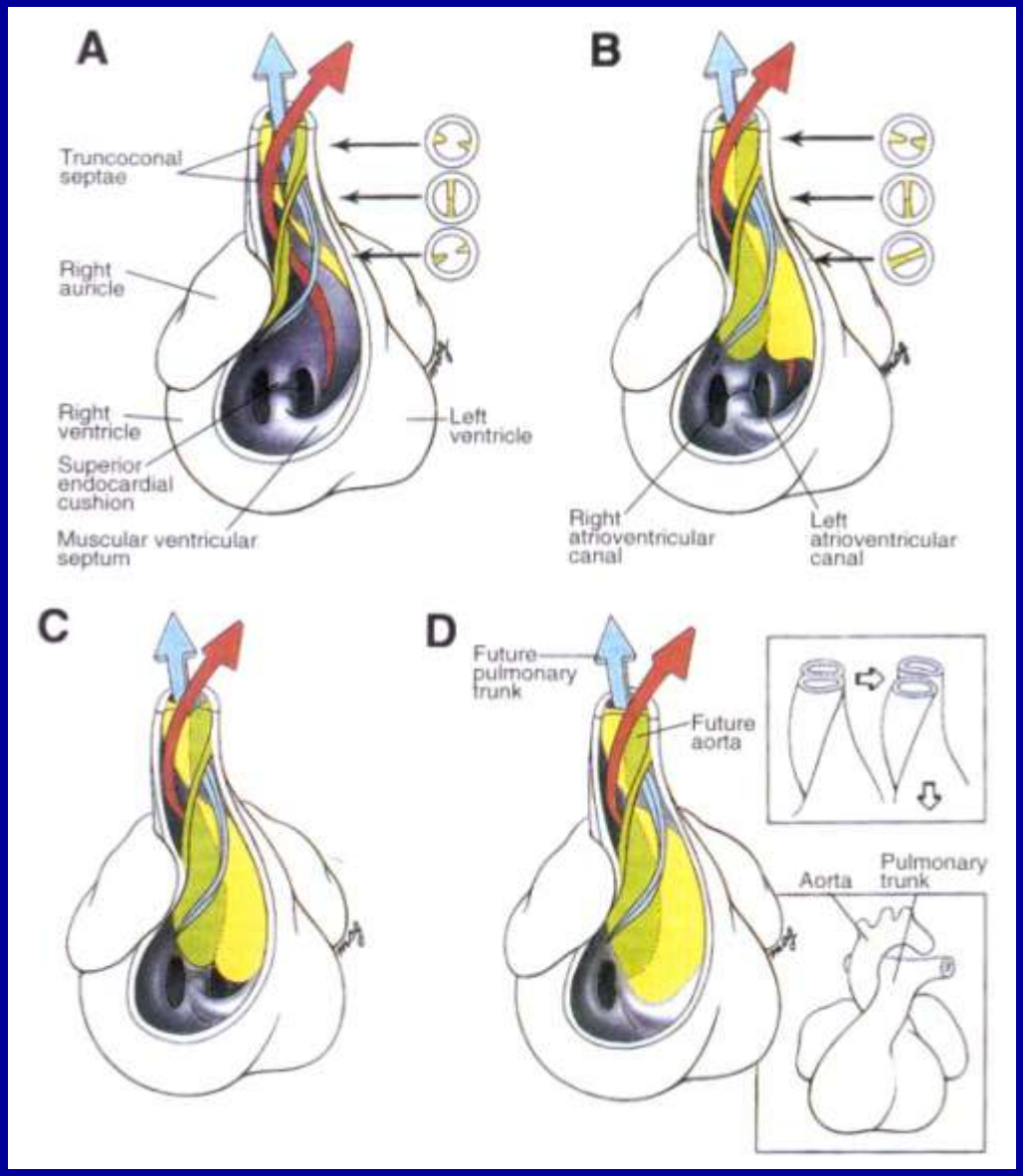
D-TGA

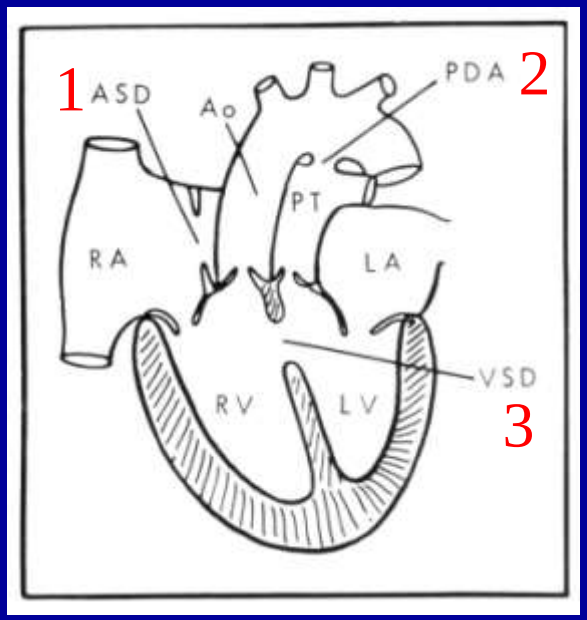
A: with intact ventricular septum and ASD and bronchial arteries
B: with VSD and no PS

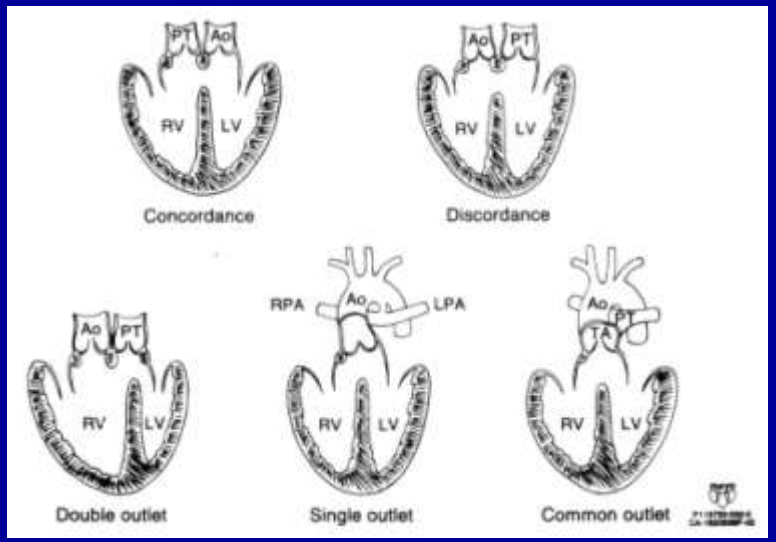


From Hurst, 1999, Ch 70

Transposition of the Great Arteries





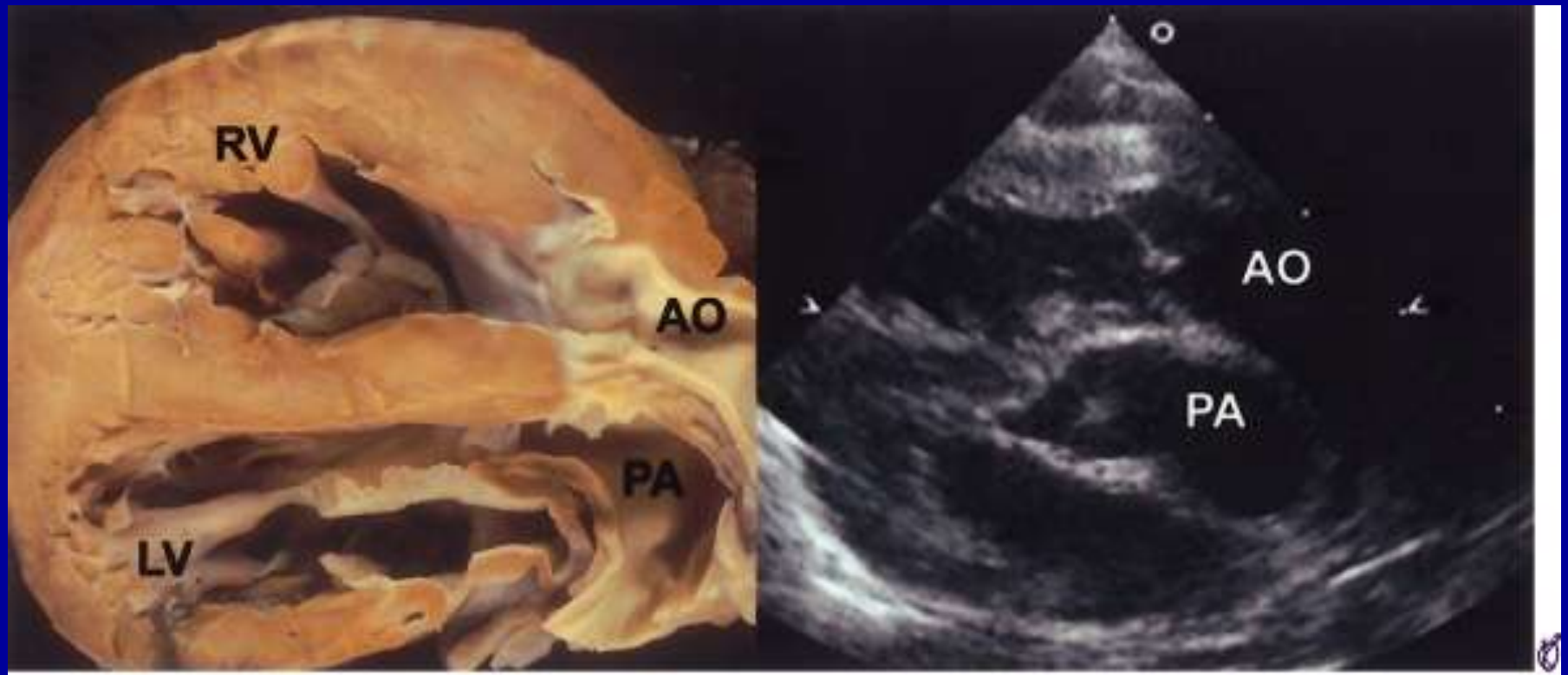


Moss and Adams, 2001, p. 14

Physiology of d-TGA

- Pulmonic circuit to systemic circuit net flow is effective systemic flow
- Systemic circuit to pulmonic circuit net flow is effective pulmonic flow
- Neonate with intact ventricular septum and closing ductus, severe hypoxemia occurs
- With large shunting sites, saturation is better, depending on vascular resistances
- Bronchopulmonary collateral circulation may help, seen in over 30% of infants

Transposition of the Great Arteries



Note the parallel nature of the aorta and pulmonary artery.

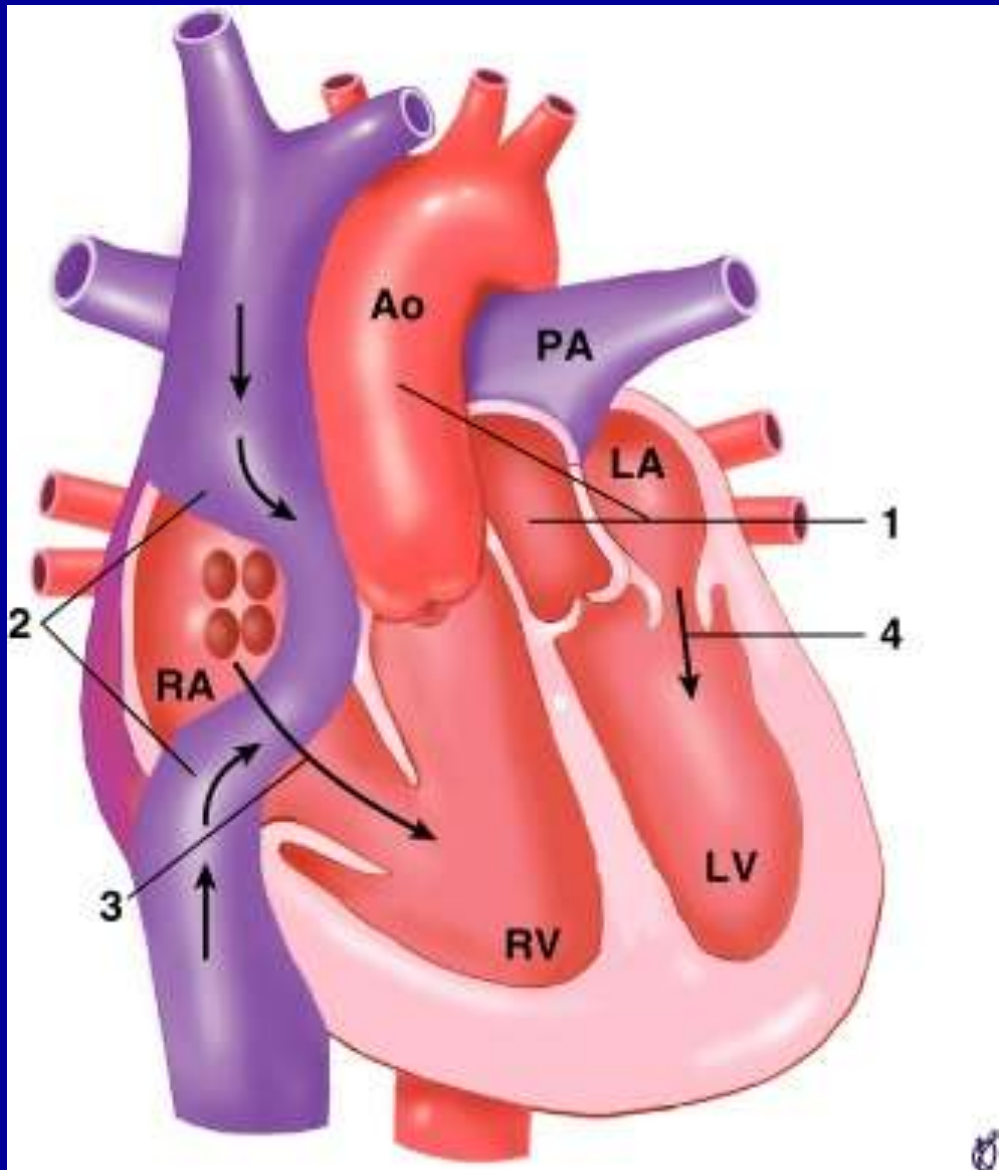
Treatment of d-TGA

- Small VSD or none: cyanosis in first hour of life
- Large VSD: CHF in 2-6 weeks
- Large VSD and LVOTO: immediate cyanosis, similar to ToF
- Formerly, cath and percutaneous balloon septostomy (“Rashkind”, very brisk procedure)
- Current, echo and complete repair of neonate

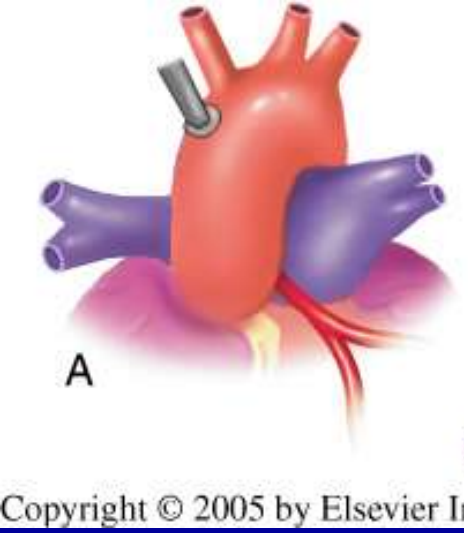
Treatment of d-TGA

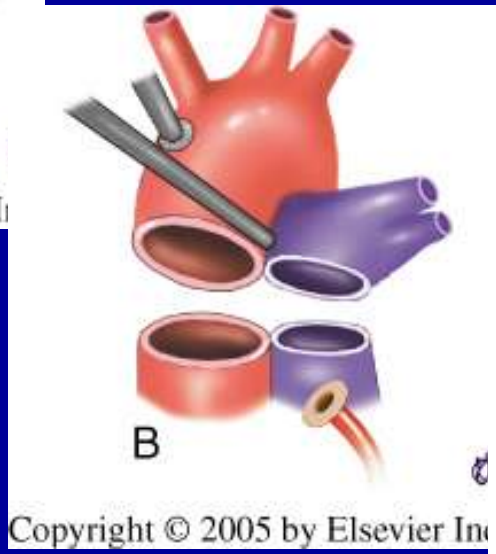
- Balloon atrial septostomy (Rashkind)
- Surgical creation of ASD (Blalock-Hanlon, needs no cardiopulmonary bypass) historical footnote
- PA banding if large VSD, formerly commonly performed
- SA-PA shunt if severe LVOTO
- Pg E1 to temporize a day or a few days
- Atrial switch, Arterial switch, VSD closure
- Rastelli procedure: LV to Ao through VSD, and RV to PA with valved conduit

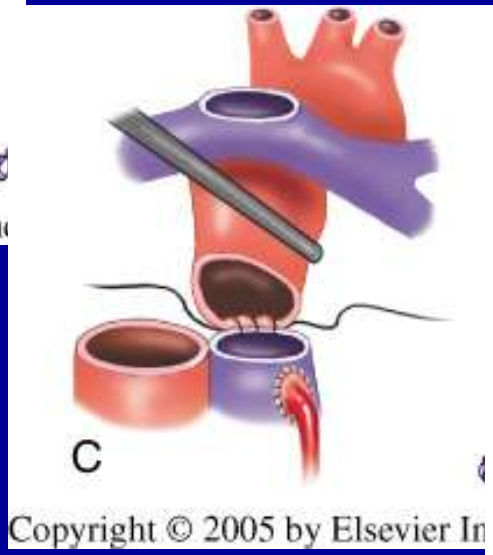
Atrial Switch (Mustard/Senning)

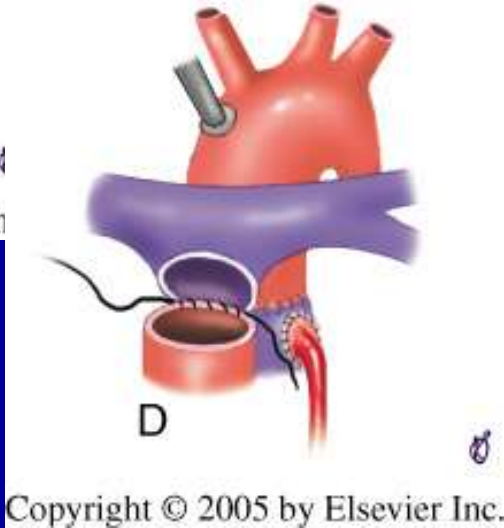


Arterial Switch (Jatene)



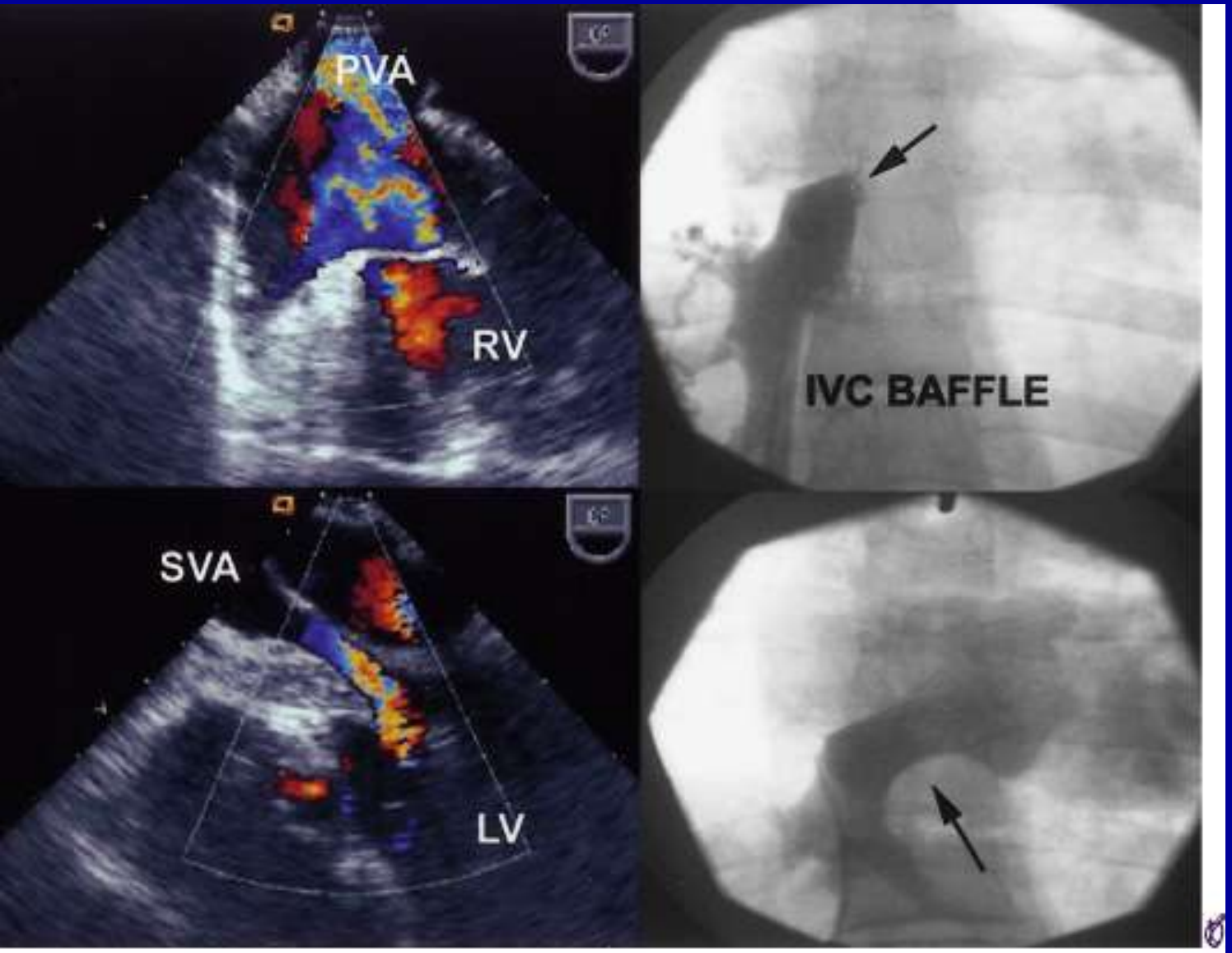






LeCompte Maneuver - The aorta is brought under the bifurcation of the pulmonary artery, and the pulmonary artery and the aorta are anastomosed without necessitating graft interposition.

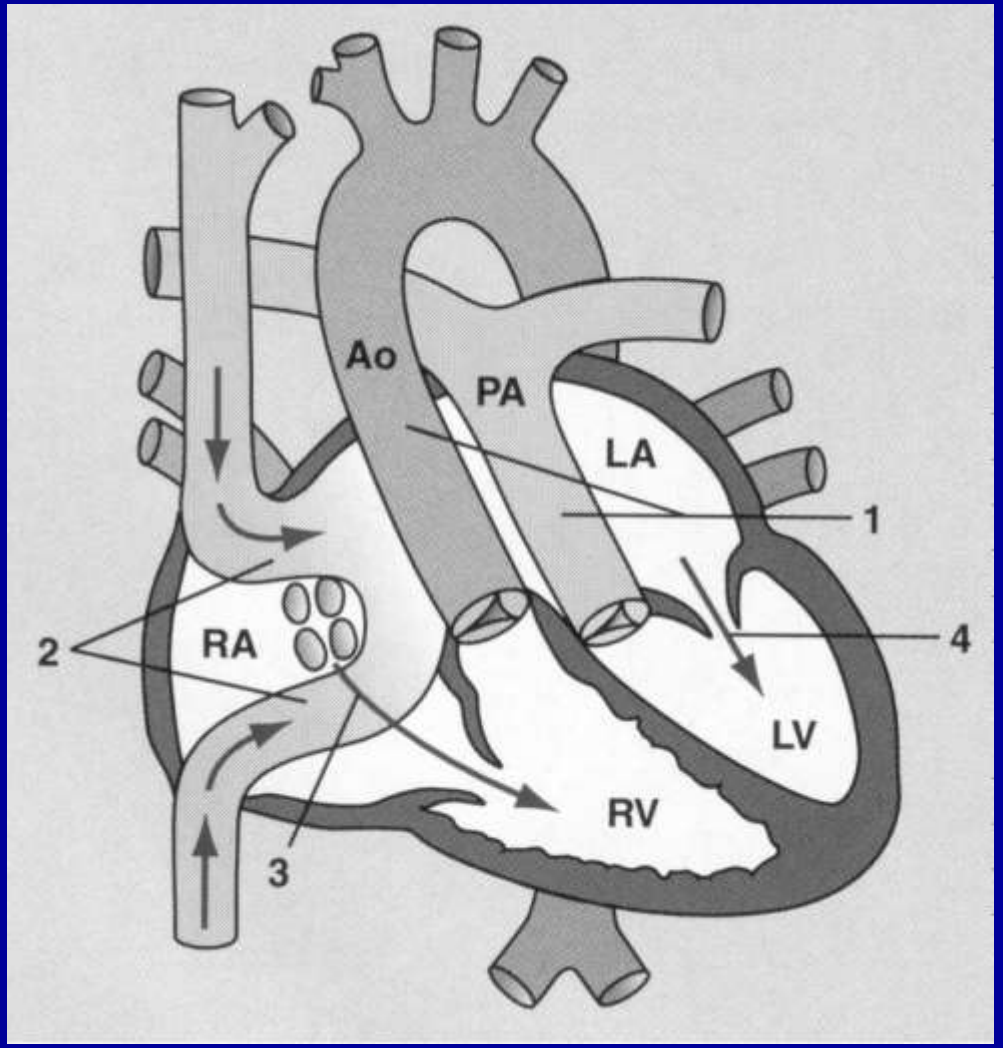
Post-Mustard Anatomy



right upper panel shows complete obstruction of the inferior limb of the systemic venous baffle, whereas the lower right panel is the same case after stenting

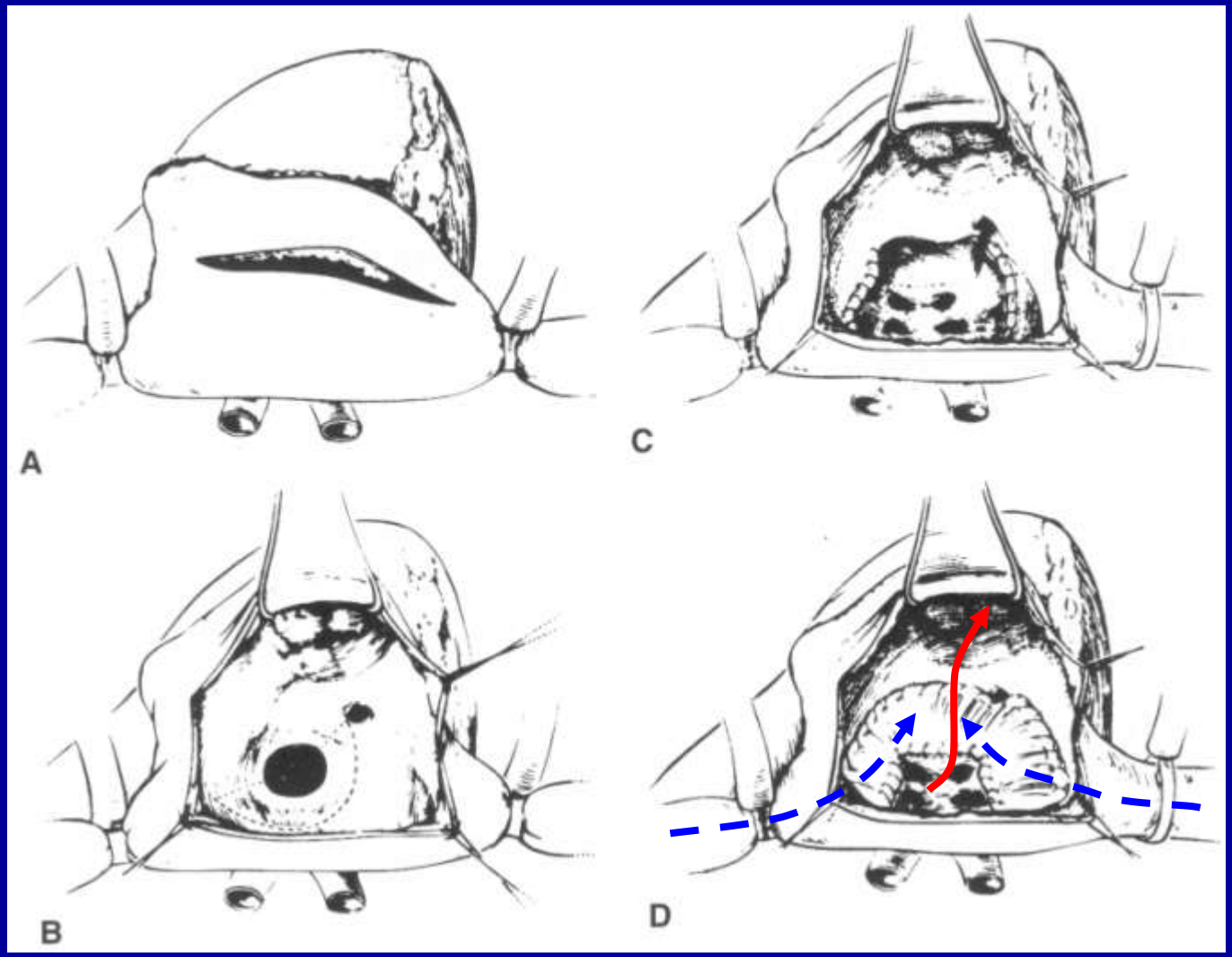
Atrial Switch

- Mustard – atrial septum is resected, pericardial baffle used
- Senning – atrial septum is baffle, blood passes over small segment of external RA free wall



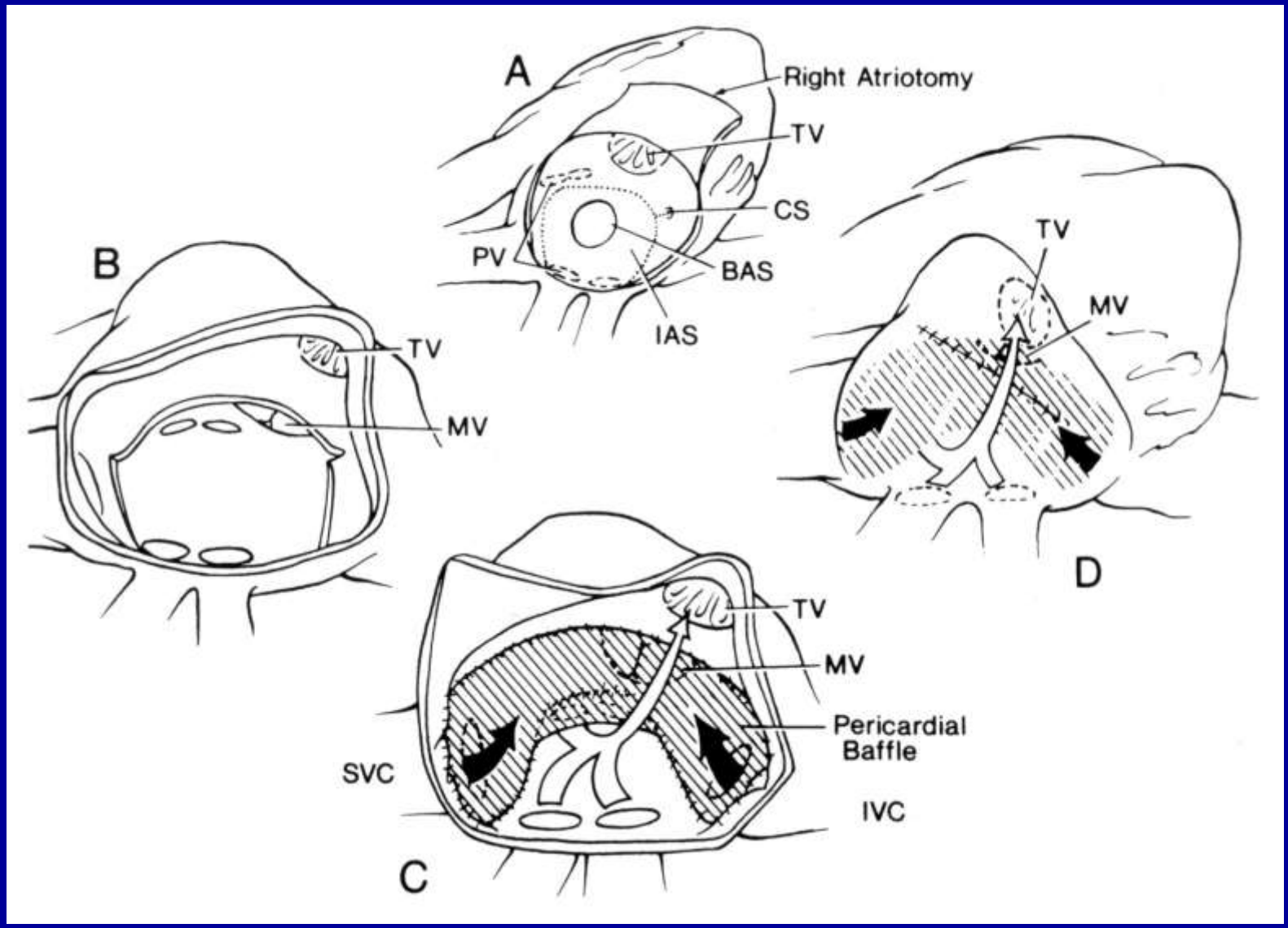
Braunwald, 2001, p. 1610

Mustard Operation



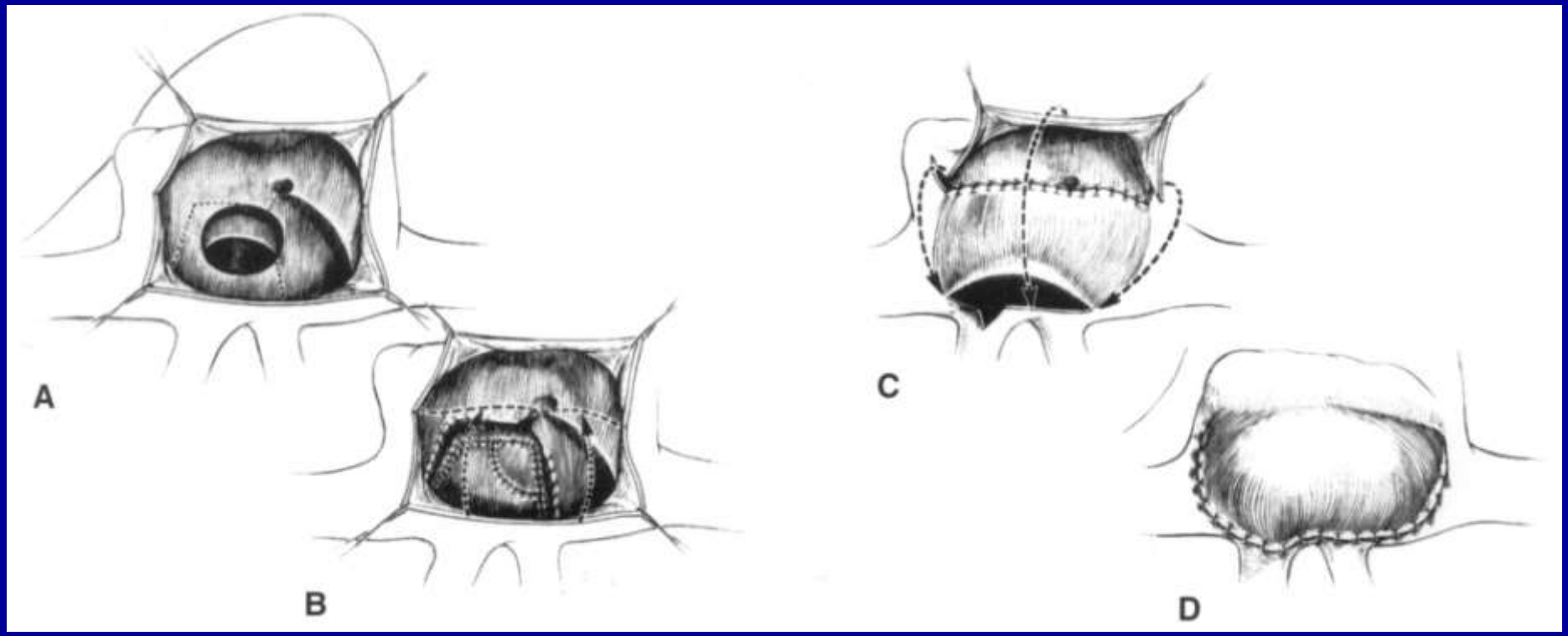
Mavroudis, 2003, p. 448

Mustard Operation



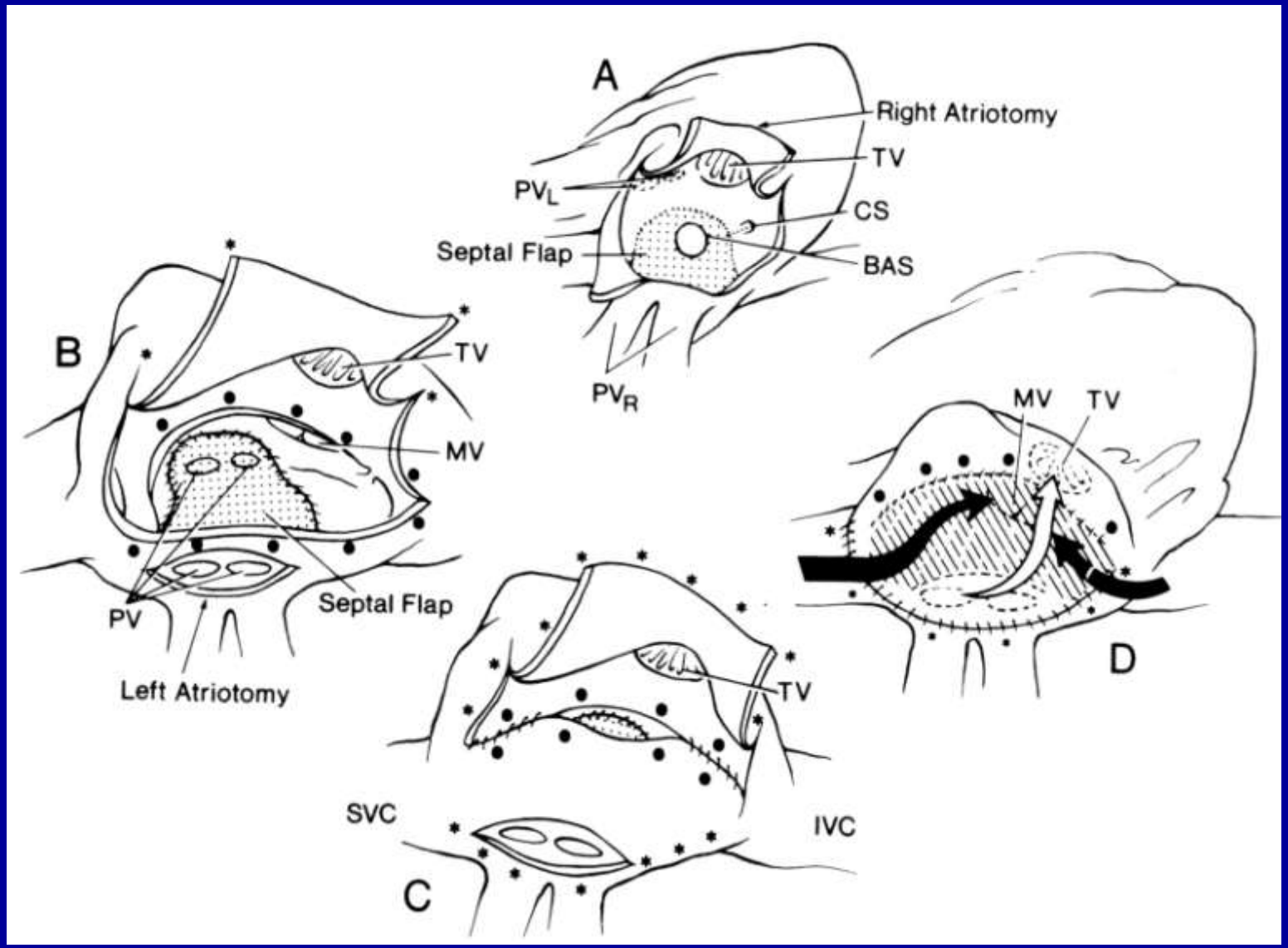
Moss and Adams, 2001, p. 1059

Senning Operation



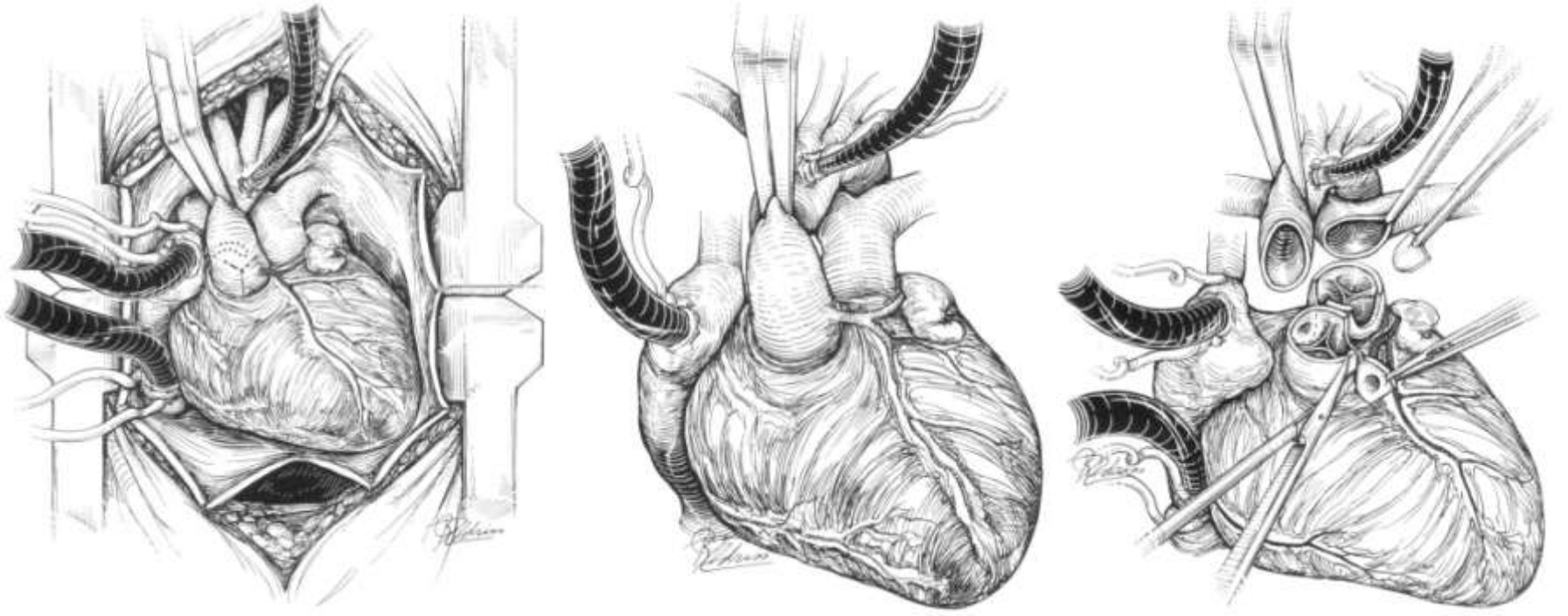
Mavroudis, 2003, p. 449

Senning Operation

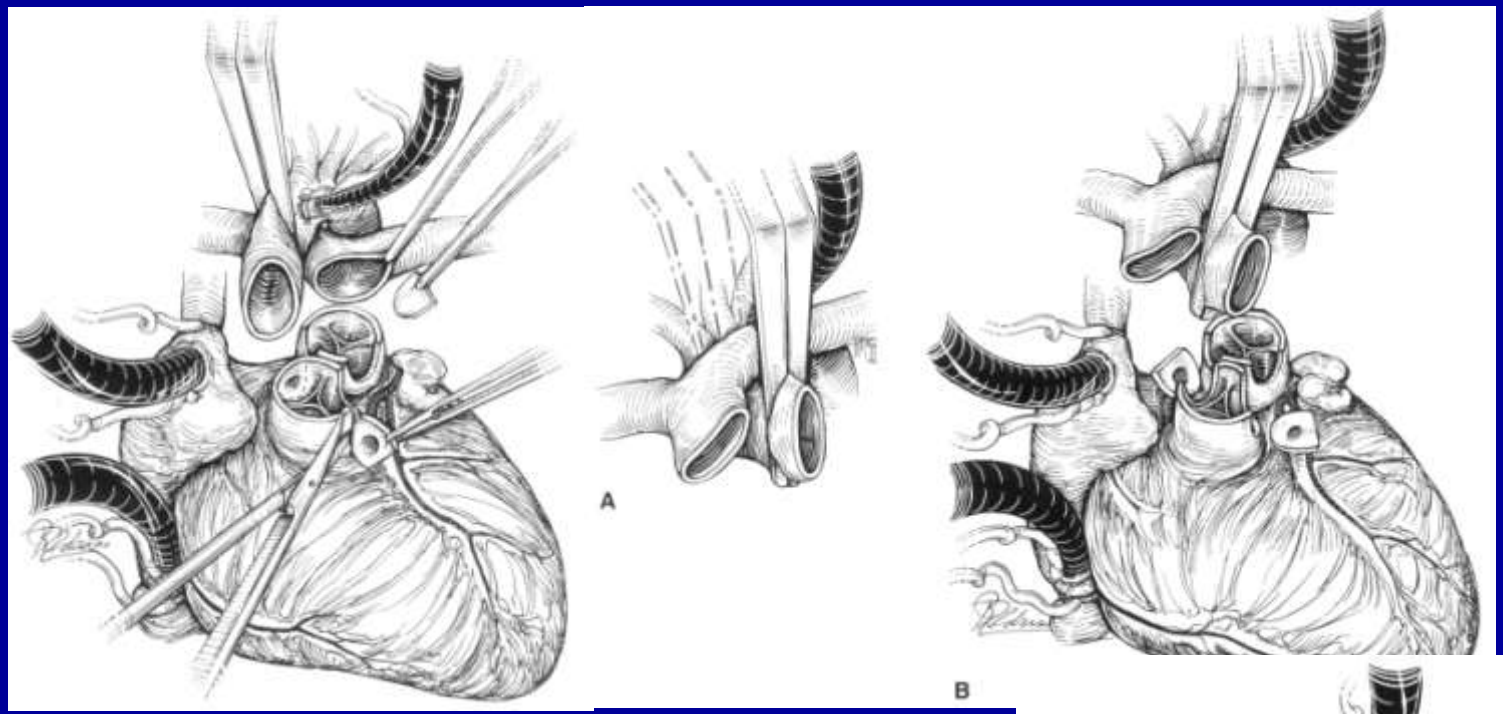


Moss and Adams, 2001, p. 1058

Arterial Switch (Jatene)



Mavroudis, 2003, p. 449



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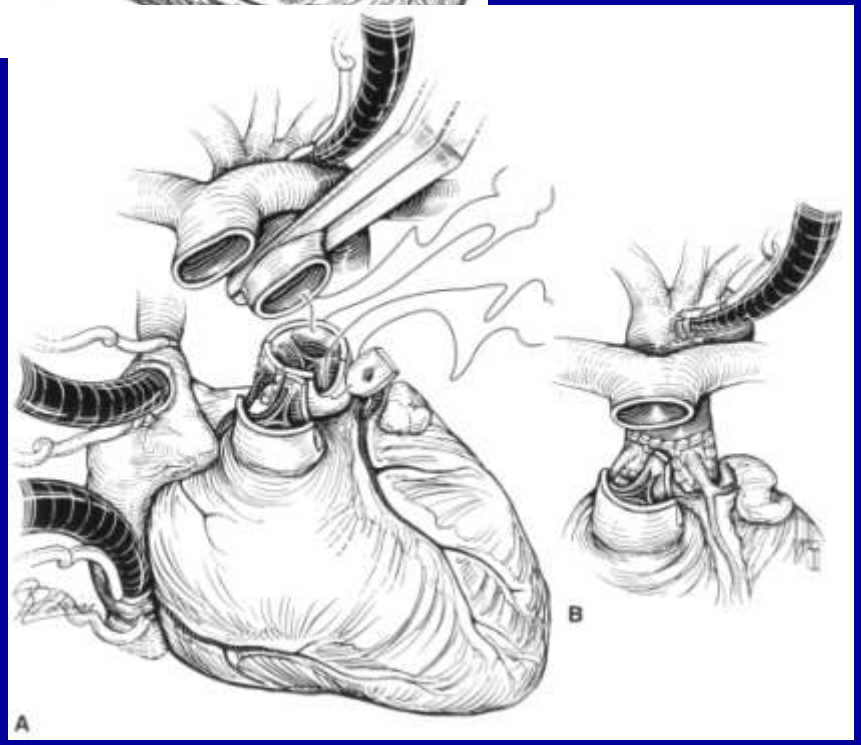
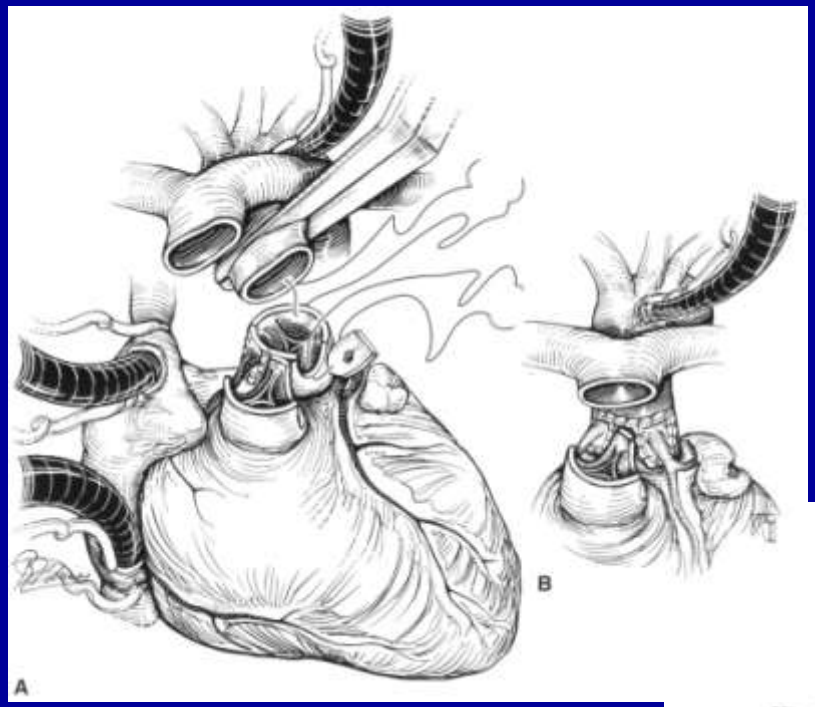
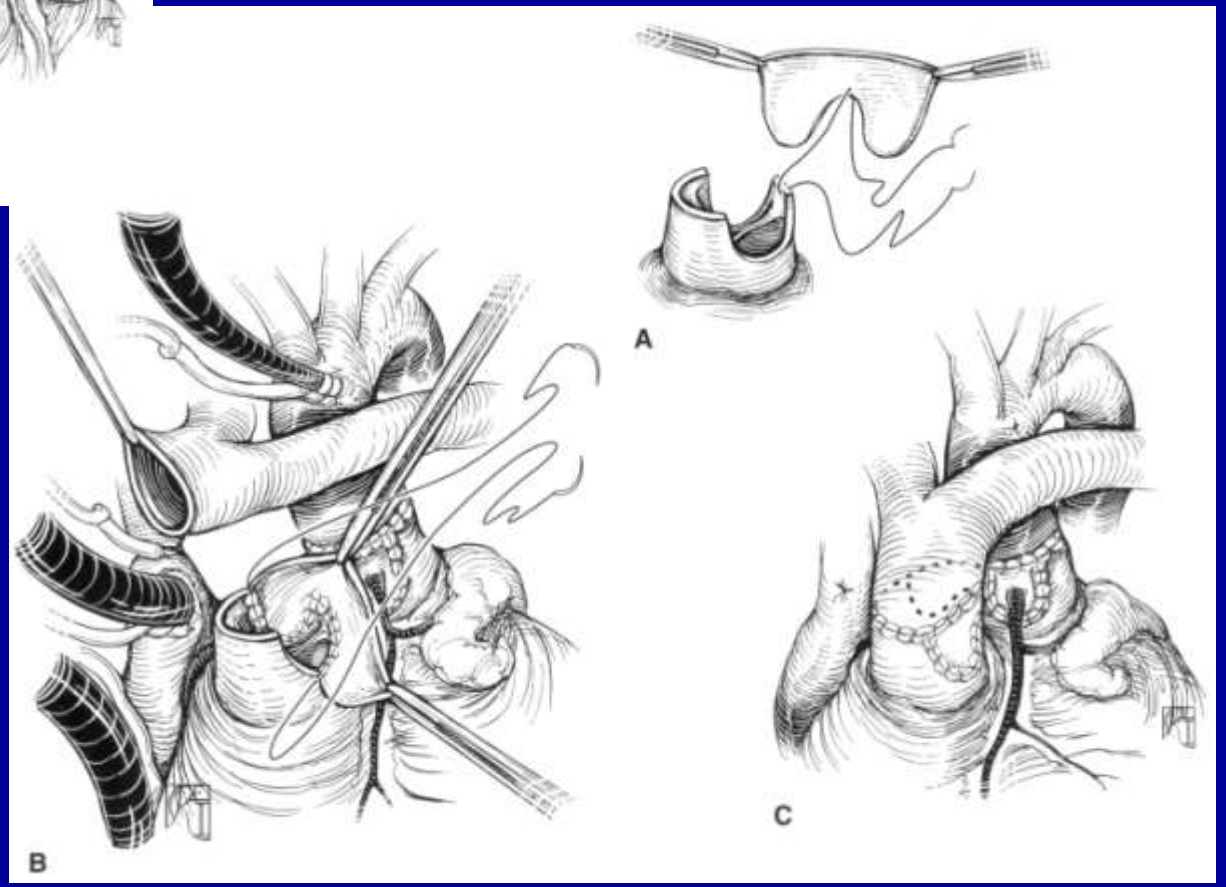


Mavroudis, 2003, p. 449

Arterial Switch (Jatene)





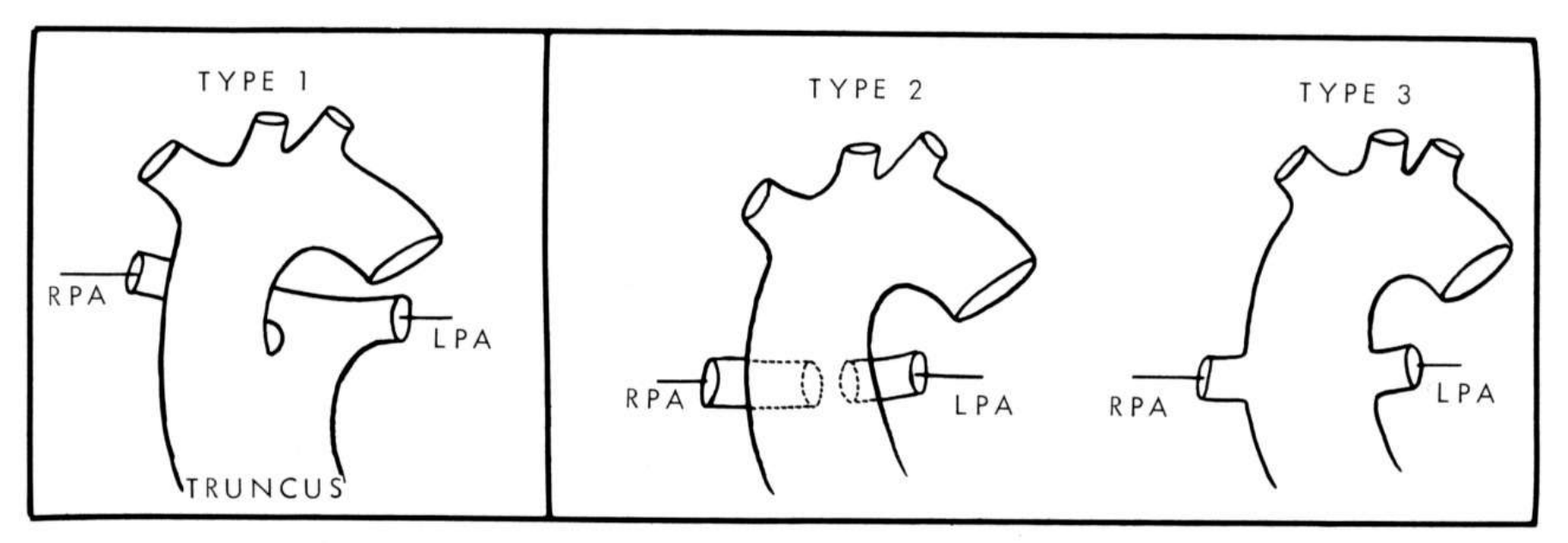
Mavroudis, 2003, p. 449

4. Truncus Arteriosus

Pathophysiology

- Definition: One truncal vessel gives rise to aortic, pulmonary and coronary circulations
- The infundibular truncal ridges fail to form
- Invariable VSD (incomplete distal pulmonary infundibulum development)
- Truncal valve – 1/3 are quadricuspid, regurg and stenosis in 10-15% each, coronary anomalies common

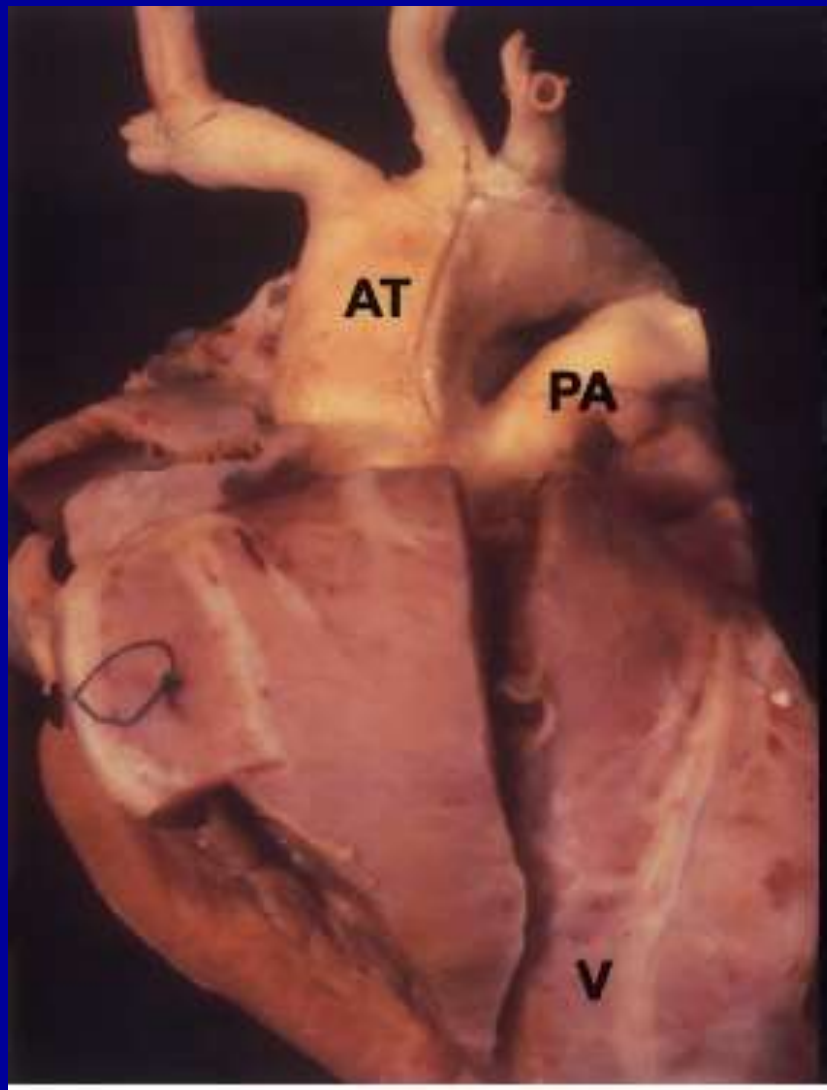
Types of Truncus Arteriosus

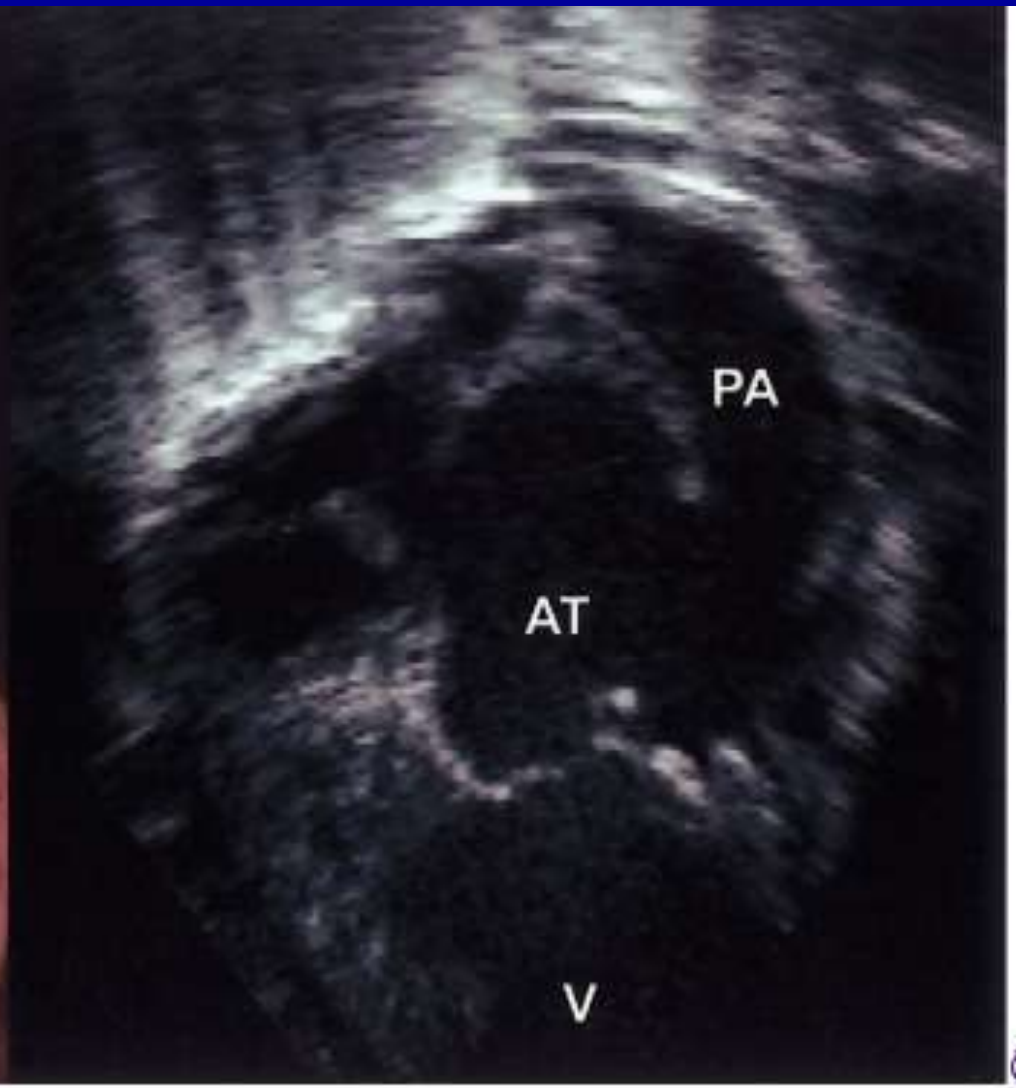


- Pulmonary flow: size of PA's and PVR
- Corrective surgery needed in first few months of life

Perloff, 1994, p. 688; Braunwald, 2001, p. 1537

Truncus Arteriosus





Surgery for Truncus Arteriosus

- Close the VSD, connected to truncal vessel
- Excise PA's from truncal vessel
- Valved conduit from RV to PA (likely will need replacement as child reaches 3-5 years)
- Address truncal valve abnormality – may be challenging
- Higher risk if severe truncal valve regurgitation, interrupted Aortic arch, coronary anomaly, or age >100 days

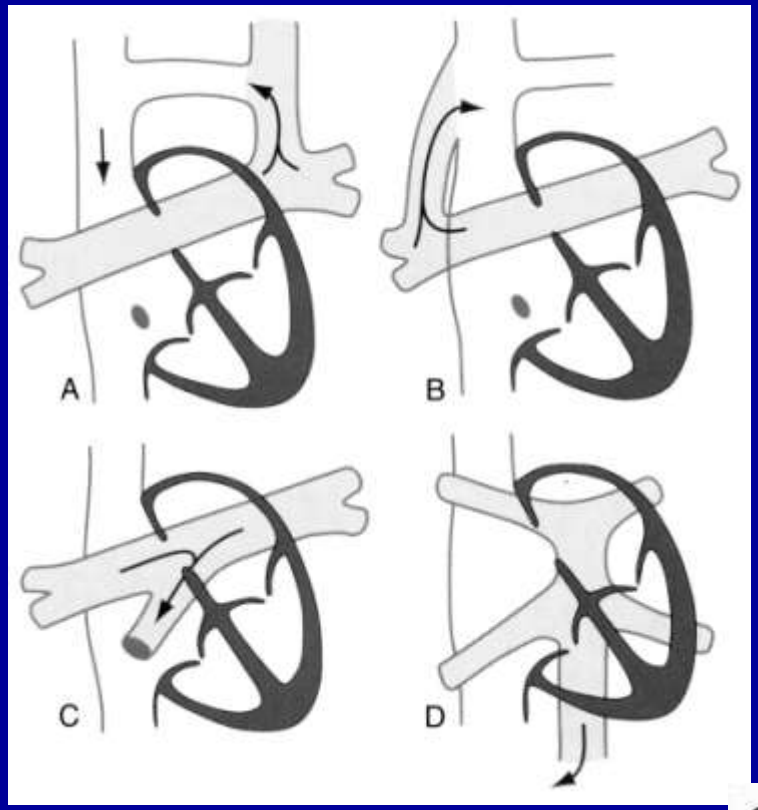
5. Total anomalous pulmonary venous return

Total Anomalous Pulmonary Venous Connection (Return)

- Cause: persistent communication of foregut plexus and cardinal or umbilicovitelline system of veins – so connection to systemic veins or RA
- Obligate ASD
- Coexistent in 30%: common atrium or atrial isomerism, single ventricle, truncus arteriosus, systemic venous anomaly
- Coexistent in 25-30%: GI, endo and GU anomalies

Braunwald, 2001, p. 1575

Total Anomalous Pulmonary Venous Connection



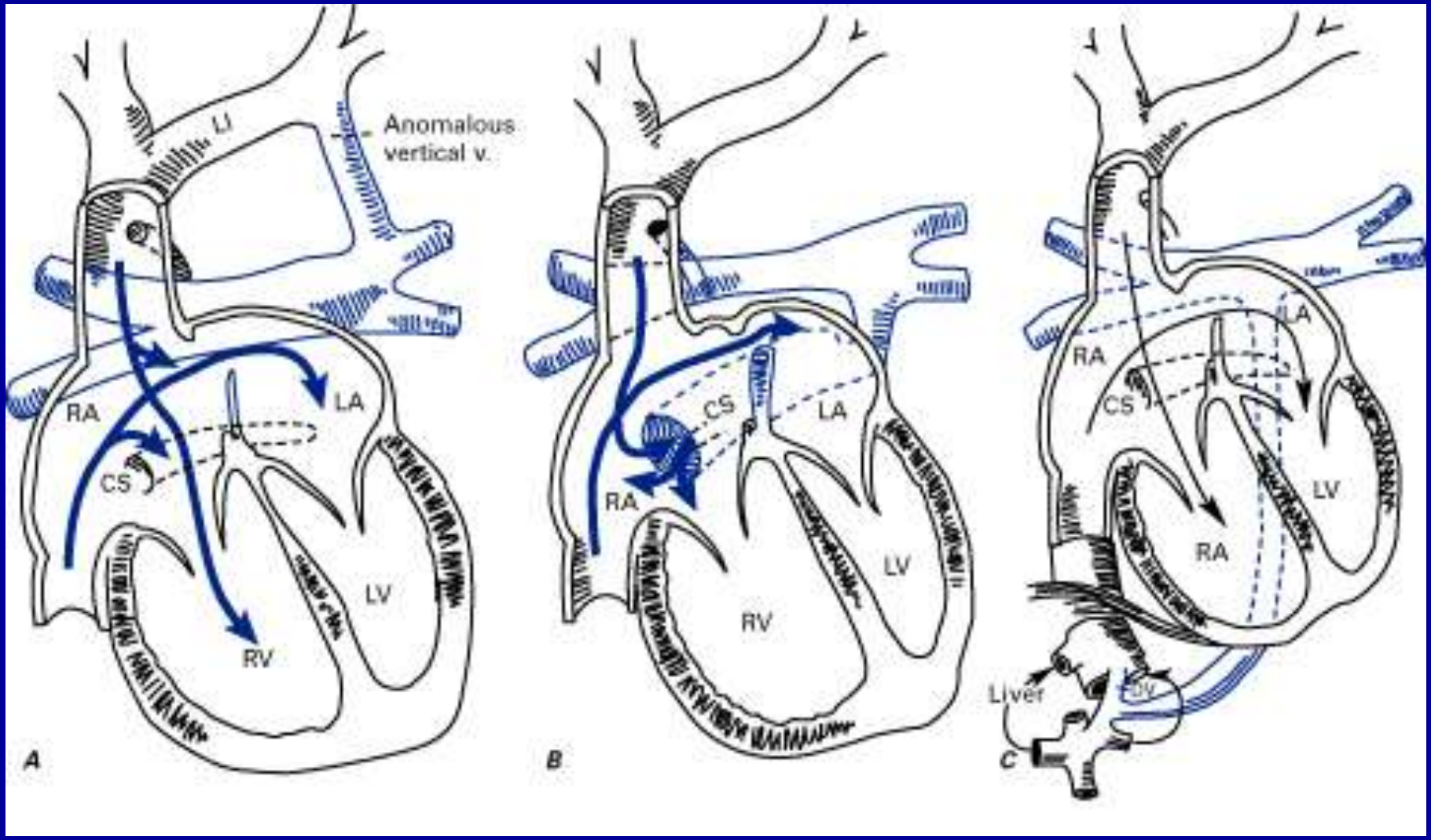
1. Above the heart
2. Into the heart
3. Below the heart (13%, more in males, obstruction and pulmonary edema)

Connection to right atrium	C.	15%
Connection to common cardinal system		
(Right) superior vena cava	B.	11%
Azygos vein		1%
Connection to left common cardinal system		
Left innominate vein	A.	36%
Coronary sinus	C.	16%
Connection to umbilicovitelline system		
Portal vein	D.	6%
Ductus venosus		4%
Inferior vena cava		2%
Hepatic vein		1%
Multiple sites		7%
Unknown		1%

Braunwald, 2001, p. 1575

TAPVR

Total anomalous pulmonary venous connection, three types:
A: to left brachiocephalic, B: to coronary sinus, C: below diaphragm

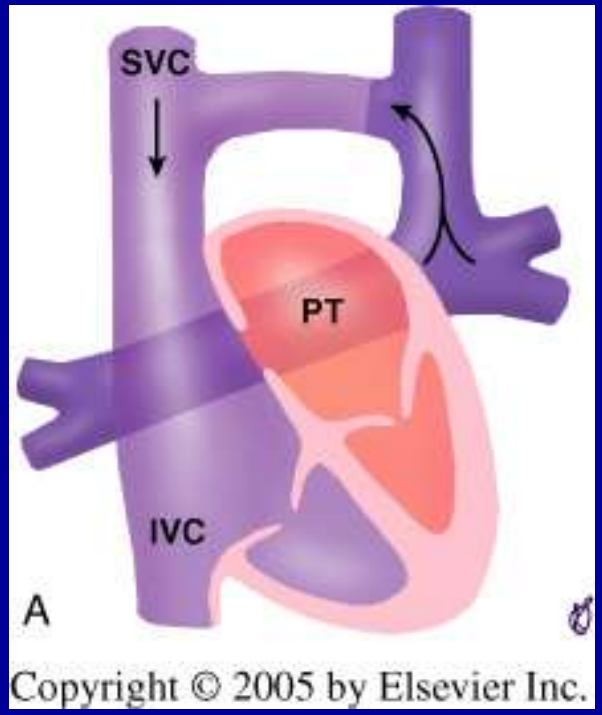


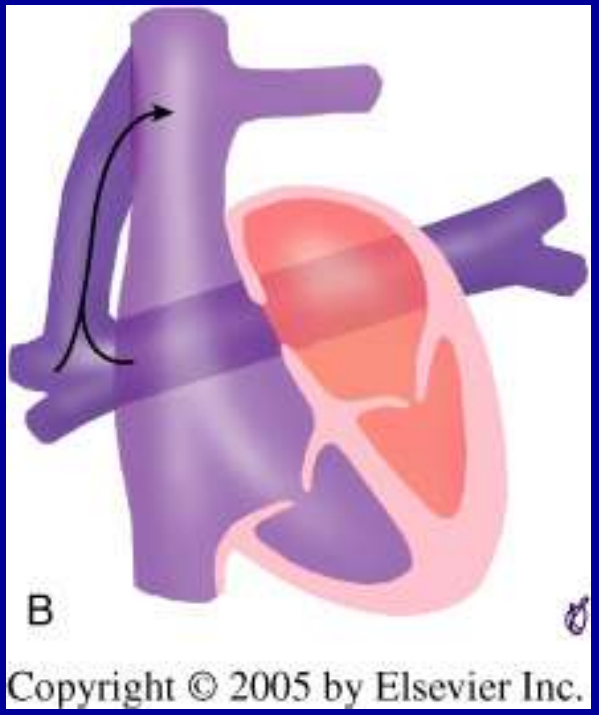
From Hurst, 1999, Ch 70

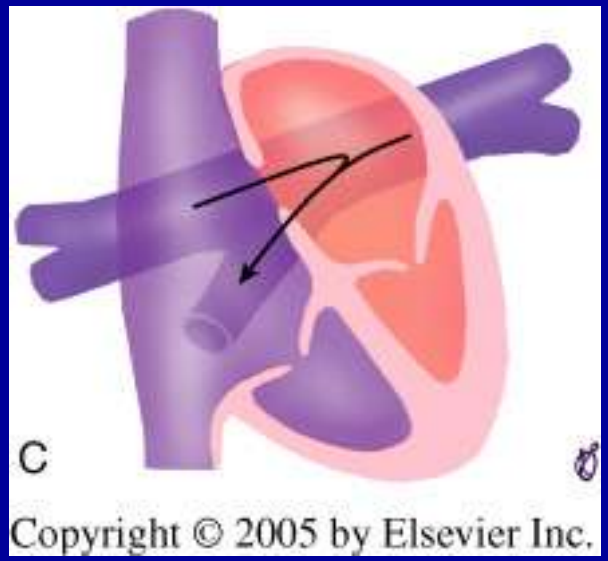
TAPVR Types

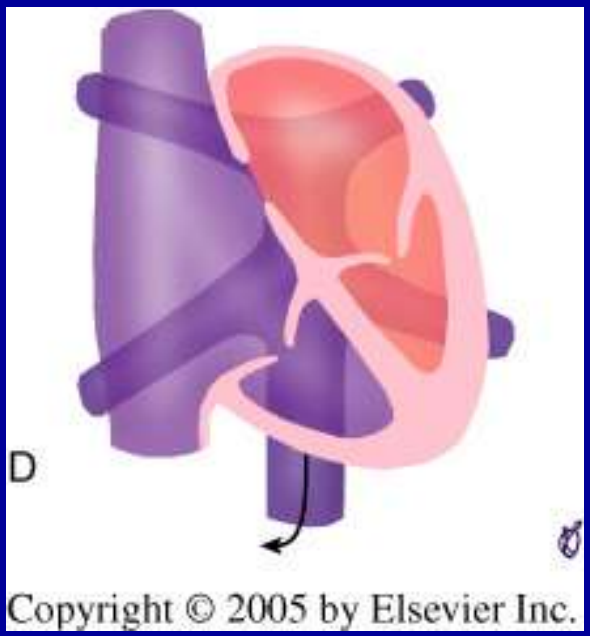
Supracardiac, in which the pulmonary veins drain either via the vertical vein to the anomalous vein **(A)** or directly to the superior vena cava (SVC) with the orifice close to the orifice of the azygos vein **(B)**.

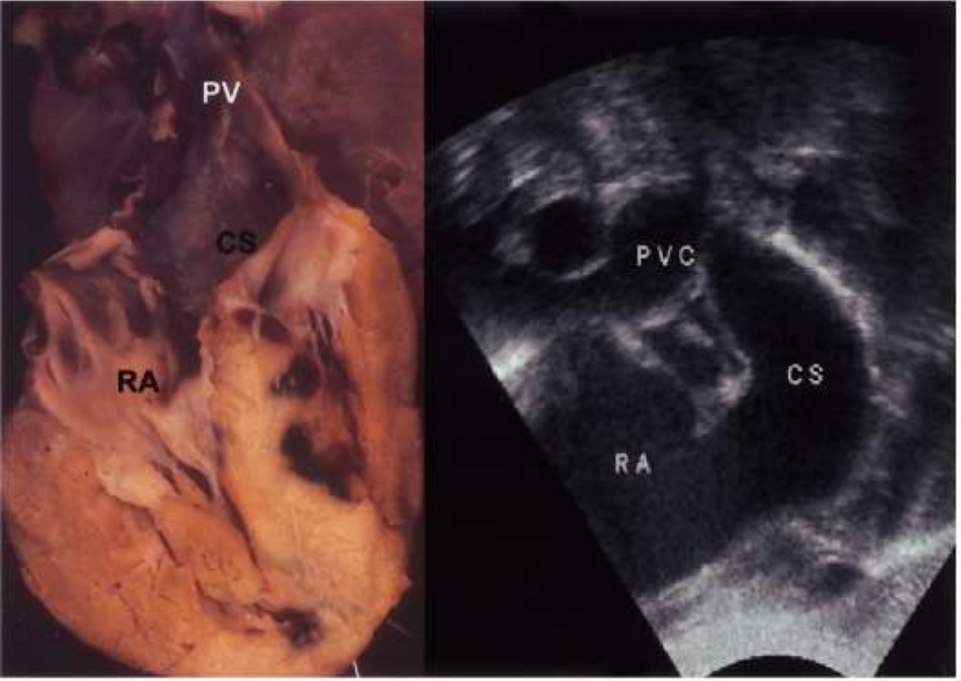
C, Drainage into the right atrium via the coronary sinus. **D,** Infracardiac drainage via a vertical vein into the portal vein or the inferior vena cava (IVC).



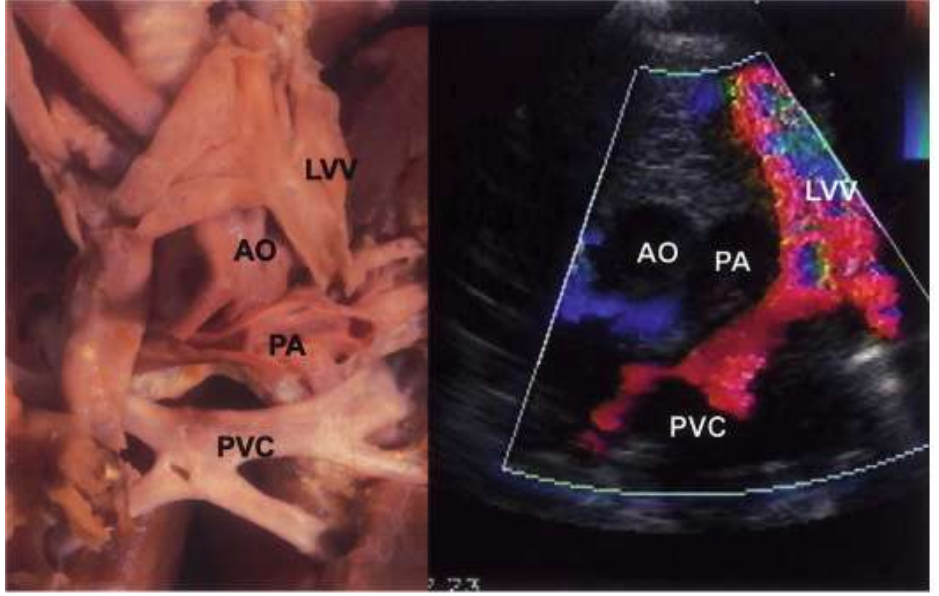






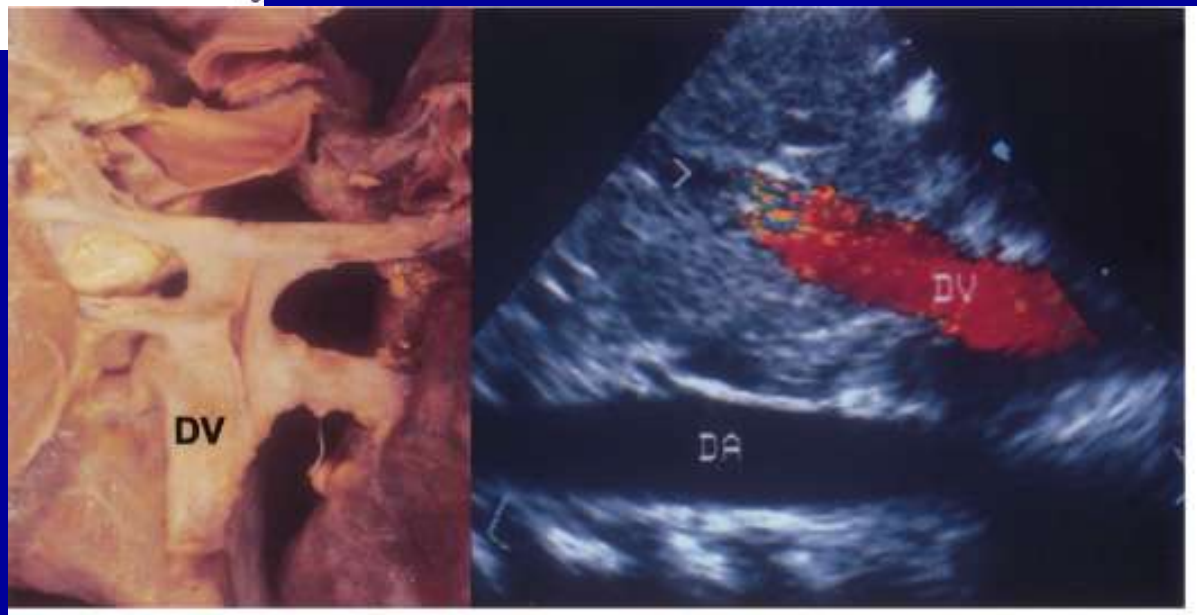


A



B

A. Subcostal view TAPVR to CS (PVC = confluence of PVs)
B. Suprasternal view TAPVR to L vertical vein
C. Subcostal view TAPVR infradiaphragm (flow is away from heart)



C

Total Anomalous Pulmonary Venous Connection

- Obstruction: pulmonary edema, systemic saturation below 70%, PA pressure more than systemic
- Unobstruction: milder cyanosis
- Management: Surgery generally in first month of life
  - Close ASD, connect anomalous veins to LA
  - Often very good result

Braunwald, 2001, p. 1576

6. Eisenmenger Physiology

Eisenmenger Physiology

- "Eisenmenger Syndrome" coined by Paul Wood: PVOD from large left to right shunt with PA pressure ~ systemic, bidirectional shunt
- From ASD, VSD, PDA, AVSD, Truncus, aortopulmonary window, univentricular heart
- Usually high PVR is established in infancy (<2 y.o.), even at birth
- Cyanosis progressive during teens and 20s
- Functional capacity decreases in 20s and after
- Survival 42% at age 25

Braunwald 2001, p. 1614

Eisenmenger Physiology

- Symptoms palpitations in 50% (atrial fibrillation/flutter 35%, VT 10%), hemoptysis 20%, PE, angina, syncope, endocarditis, CHF
- Eisenmenger PDA can have pink right nail beds and cyanosis of left hand
- Management: flu shots, iron replacement, antiarrhythmics, dig, diuretics, bedrest for hemoptysis
- Lung transplant and repair of defect, or heart-lung
- General anesthesia for noncardiac surgery is high-risk, try local; paradoxic emboli

Braunwald 2001, p. 1614

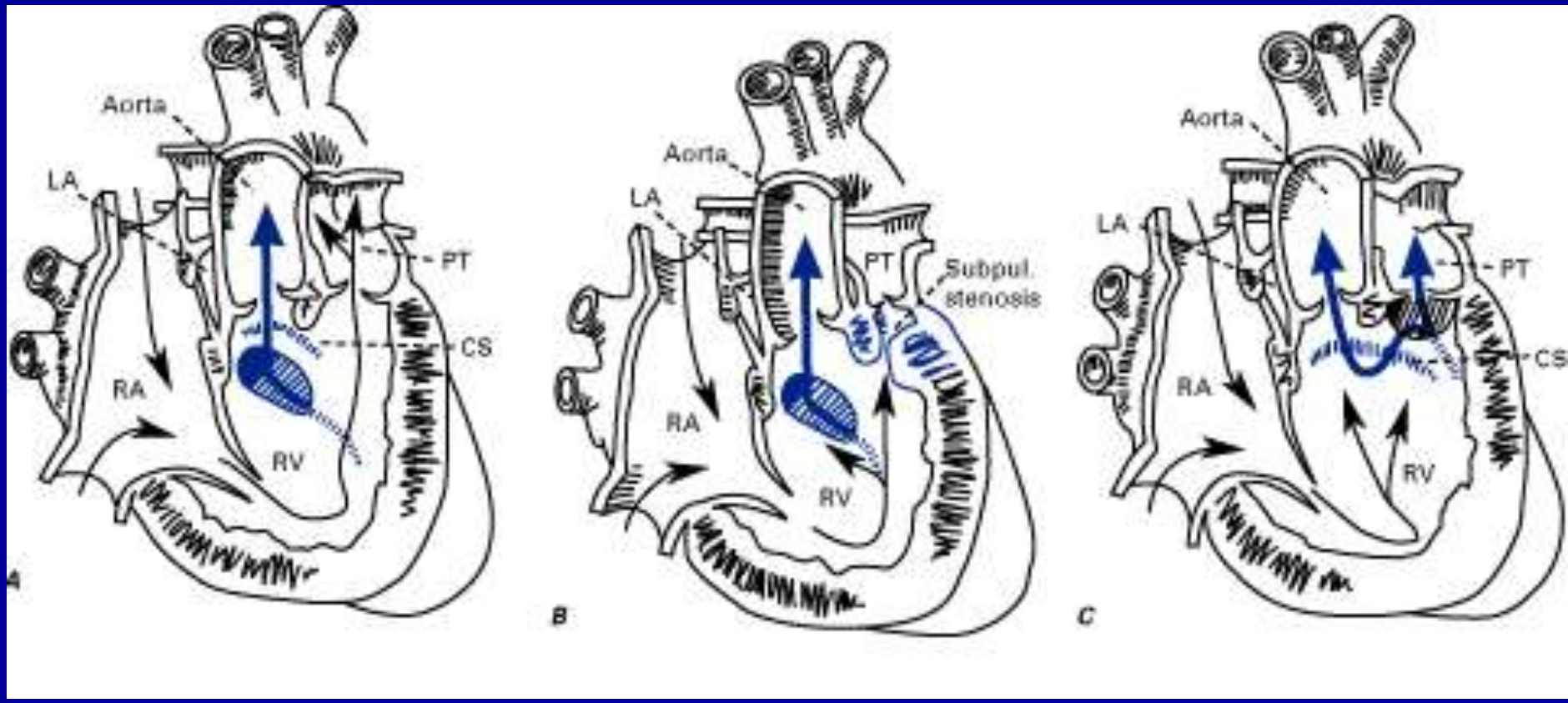
7. Complex Disease

Complex Congenital Heart Disease

- Left-right abnormalities
  - Situs inversus
  - Situs solitus
  - Combinations
  - Left isomerism (polysplenia)
  - Right isomerism (asplenia)

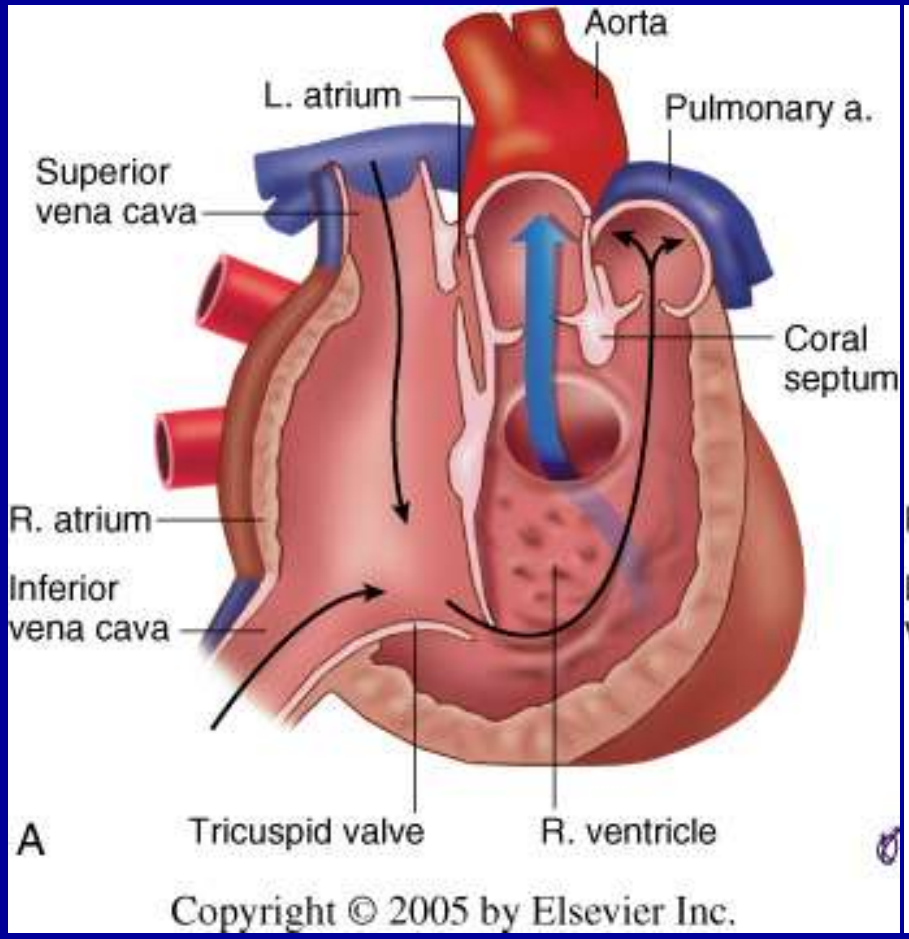
Double Outlet RV

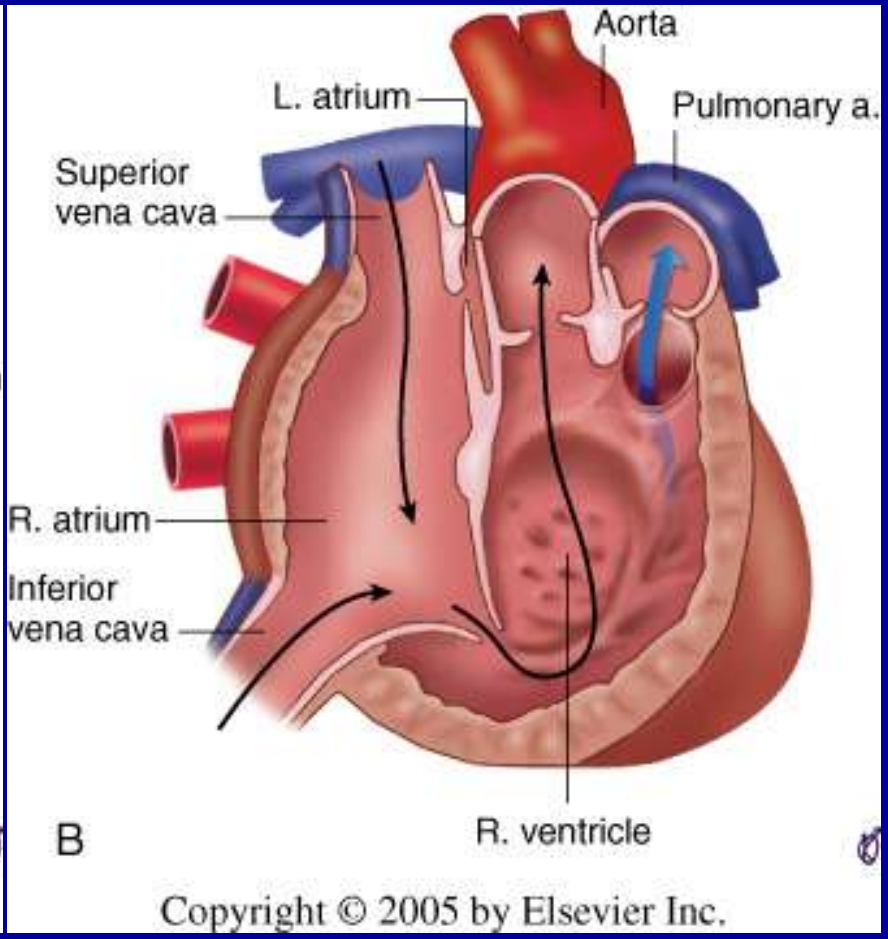
A: with subaortic VSD and no PS
B: with subaortic VSD and subpulmonary stenosis
C: with subpulmonary, supracristal VSD (Taussig-Bing complex)



From Hurst, 1999, Ch 70

Double-outlet RV

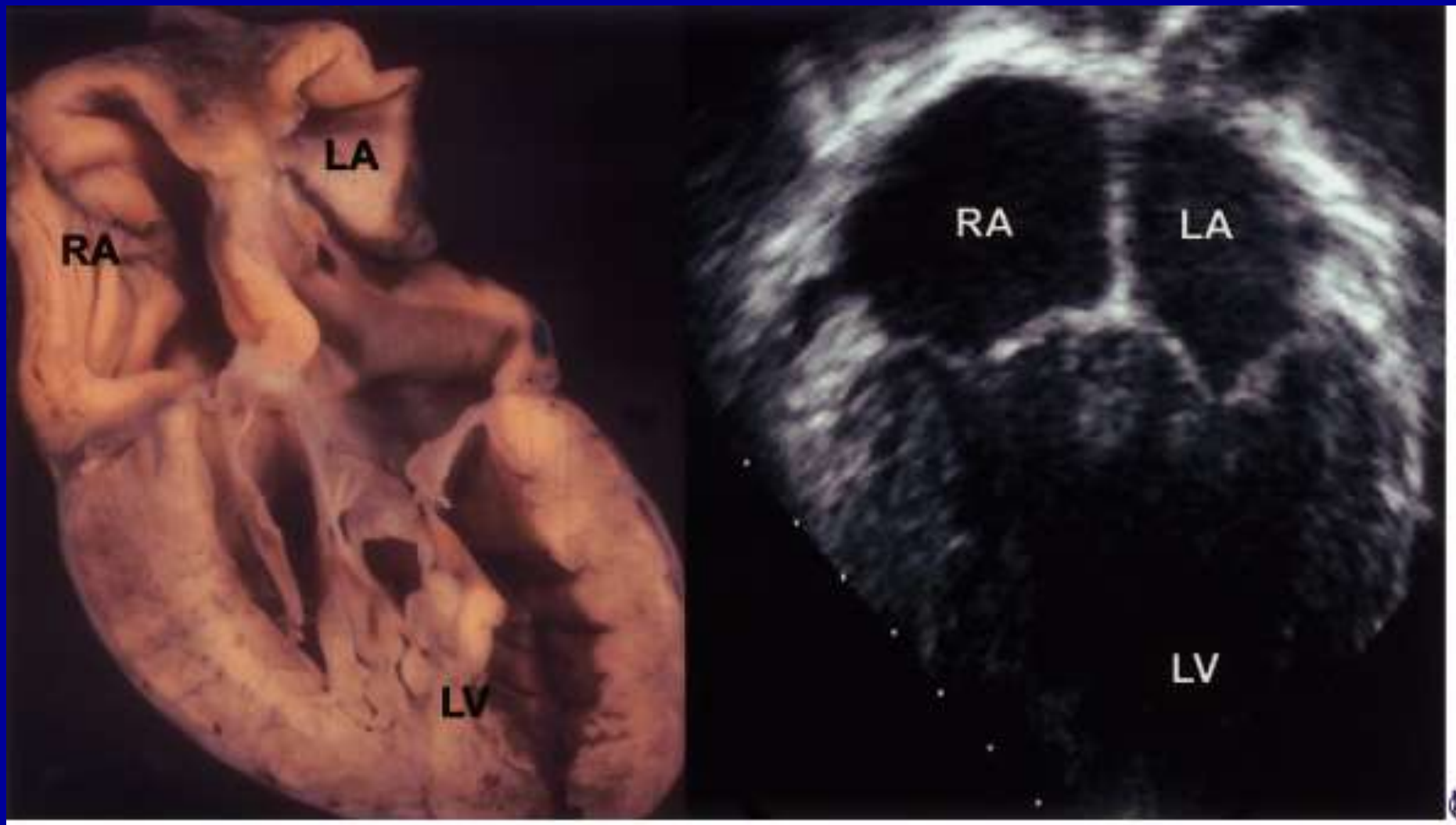




A subaortic ventricular septal defect below the crista supraventricularis favors delivery of left ventricular blood to the aorta

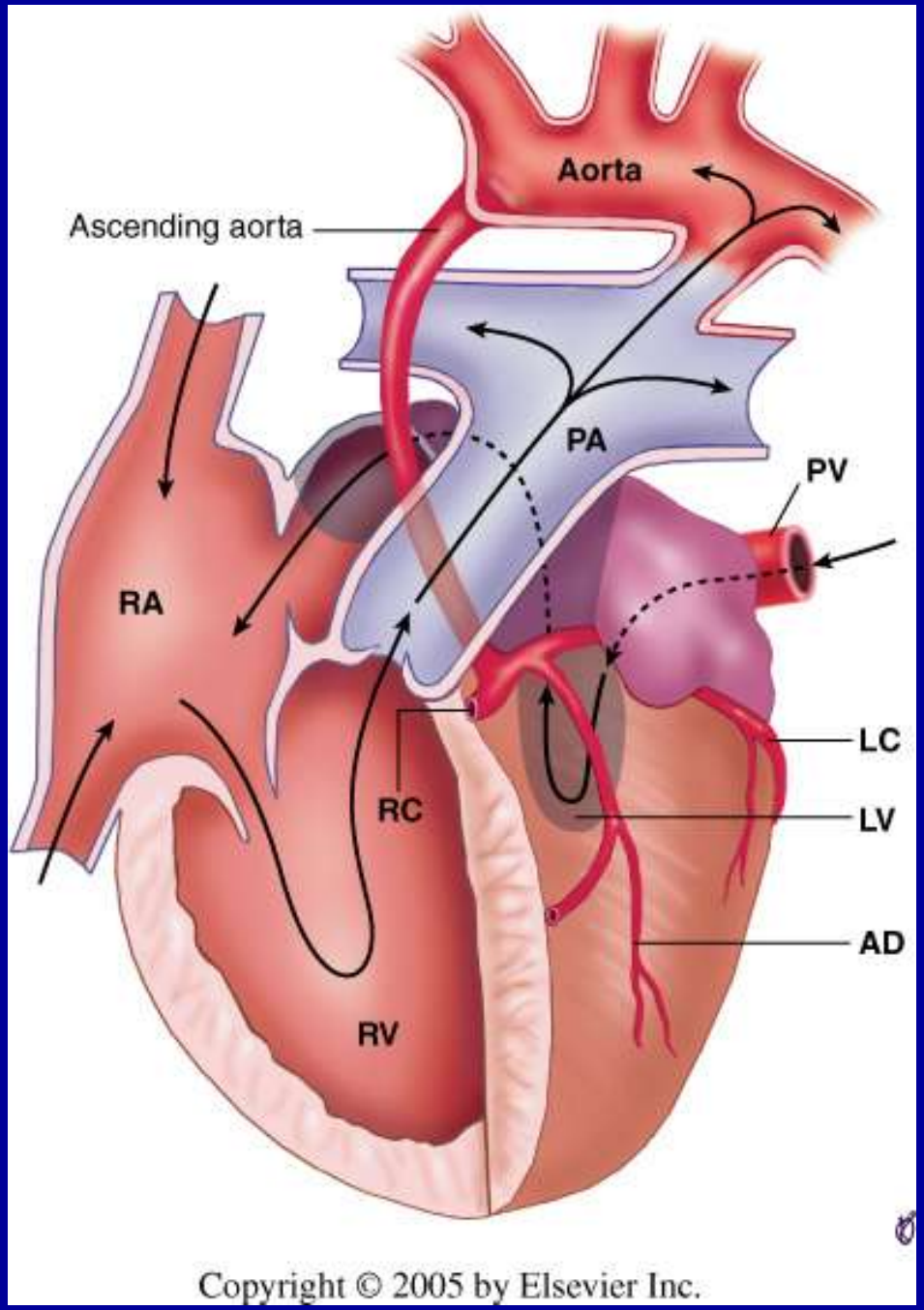
Subpulmonary location of the ventricular septal defect above the crista favors streaming to the pulmonary trunk.

Double-inlet univentricular connection of LV type (DILV)

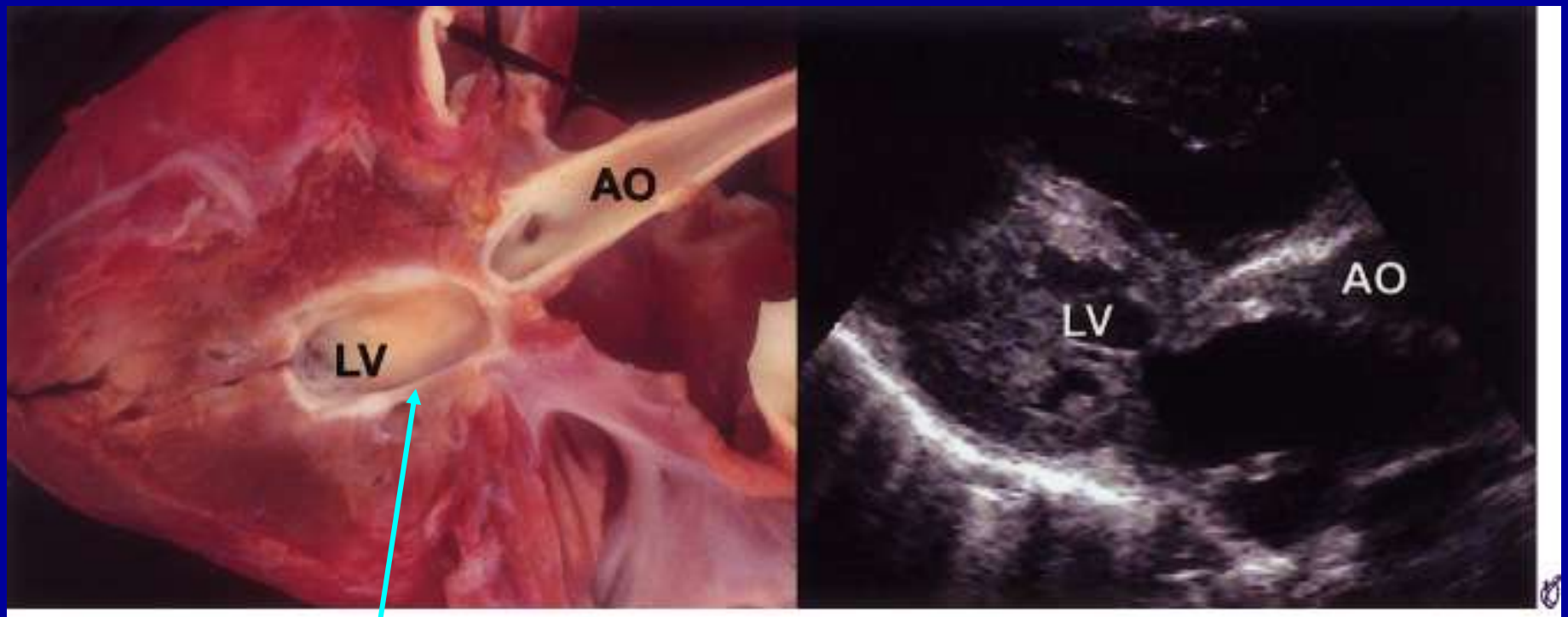


Hypoplastic Left Heart

Aortic hypoplasia, aortic valve atresia, and a hypoplastic mitral valve and left ventricle



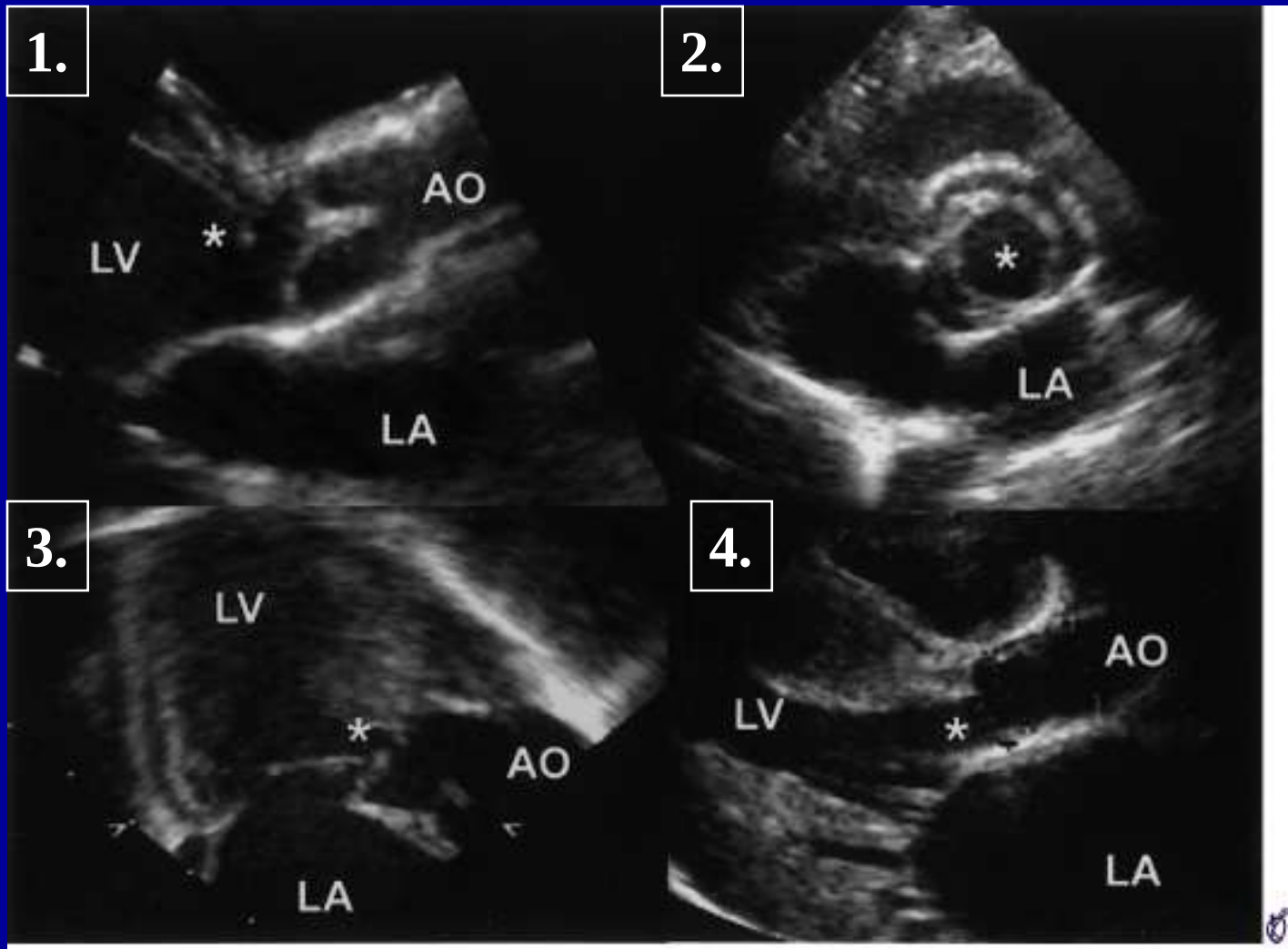
Hypoplastic Left Heart Syndrome



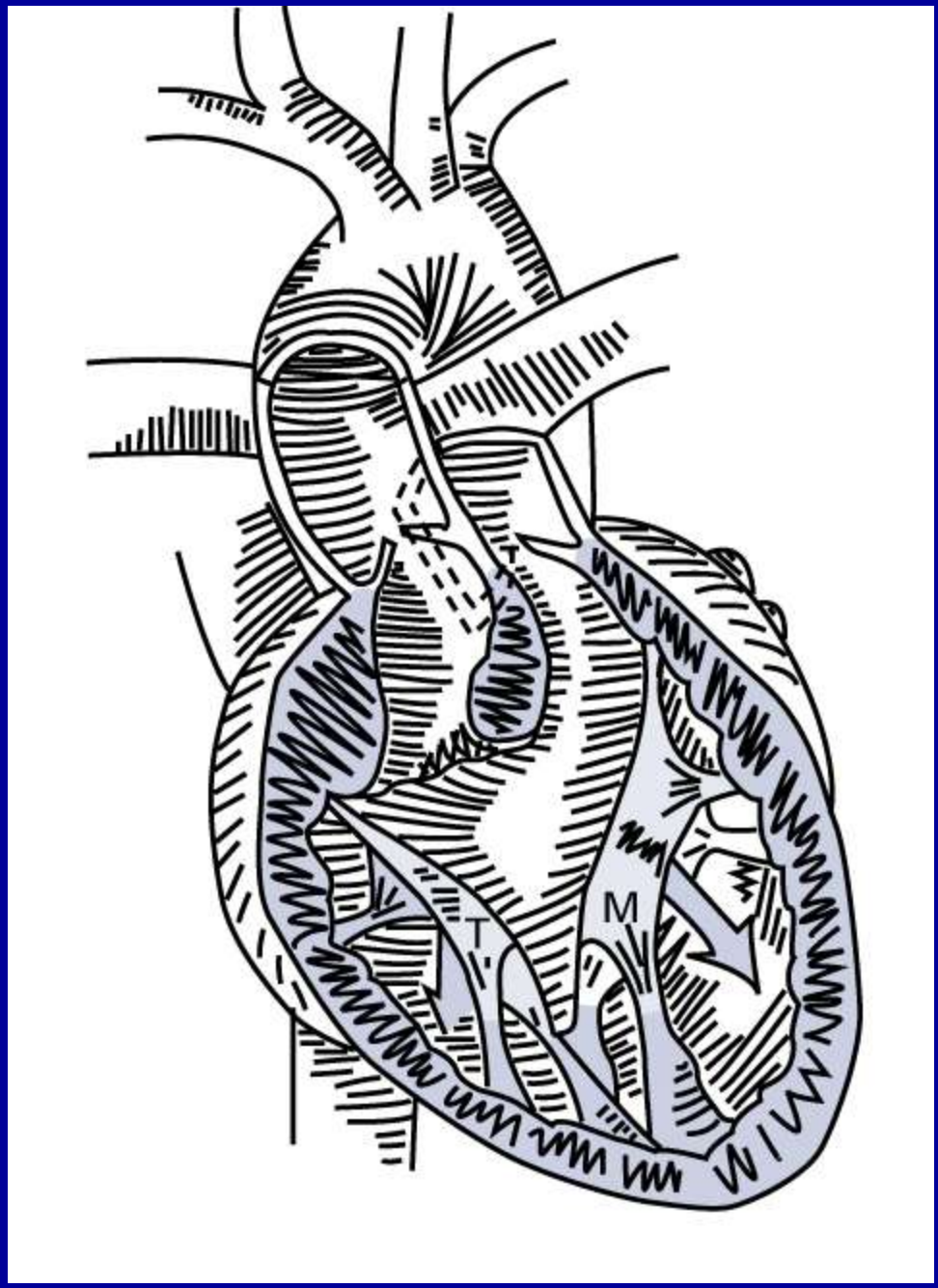
Note the associated endocardial fibroelastosis

Types of LVOT Obstruction

1. Isolated fibromuscular obstruction
2. Bicuspid Aortic valve
3. Anterior MV leaflet chordal apparatus
4. Tunnel narrowing of valve, annulus and subvalve level



Common ventricle with dextro malposition and no PS



From Hurst, 1999, Ch 70

Approach to Echocardiography in Complex Congenital Heart Disease

- Segmental Analysis
- Systemic vein to atrium connection
- Pulmonary vein to atrium connection
- Location of RA, location of LA
- Atrioventricular connections
- Location of RV, location of LV
- Ventriculoarterial connection

Differentiation of Left and Right

- Left atrial appendage is narrower and longer than right
- TV is always with RV, and MV with LV
- TV connects to IVS apically from the MV
- RV has moderator band and heavier trabeculae than LV
- Ao V and MV fibrous continuity usually

TABLE 56–1 Types of Adult Patients with Simple Congenital Heart Disease*

Native disease
Isolated congenital aortic valve disease
Isolated congenital mitral valve disease (except parachute valve, cleft leaflet)
Isolated patent foramen ovale or small atrial septal defect
Isolated small ventricular septal defect (no associated lesions)
Mild pulmonic stenosis
Repaired conditions
Previously ligated or occluded ductus arteriosus
Repaired secundum or sinus venosus atrial septal defect without residua
Repaired ventricular septal defect without residua

From Webb G, Williams R, Alpert J, et al: 32nd Bethesda Conference: Care of the Adult with Congenital Heart Disease, October 2-3, 2000. J Am Coll Cardiol 37:1161-1198, 2001.

*These patients can usually be cared for in the general medical community.

TABLE 56–2	Types of Adult Patients with Congenital Heart Disease of Moderate Severity*
Aorto–left ventricular fistulas	
Anomalous pulmonary venous drainage, partial or total	
Atrioventricular septal defects (partial or complete)	
Coarctation of the aorta	
Ebstein anomaly	
Infundibular right ventricular outflow obstruction of significance	
Ostium primum atrial septal defect	
Patent ductus arteriosus (not closed)	
Pulmonary valve regurgitation (moderate to severe)	
Pulmonic valve stenosis (moderate to severe)	
Sinus of Valsalva fistula/aneurysm	
Sinus venosus atrial septal defect	
Subvalvular or supravalvular aortic stenosis (except HOCM)	
Tetralogy of Fallot	
Ventricular septal defect with Absent valve or valves Aortic regurgitation Coarctation of the aorta Mitral disease Right ventricular outflow tract obstruction Straddling tricuspid/mitral valve Subaortic stenosis	

HOCM = hypertrophic obstructive cardiomyopathy.

From Webb G, Williams R, Alpert J, et al: 32nd Bethesda Conference: Care of the Adult with Congenital Heart Disease, October 2-3, 2000. J Am Coll Cardiol 37:1161-1198, 2001.

*These patients should be seen periodically at regional adult congenital heart disease centers.

TABLE 56–3 Types of Adult Patients with Congenital Heart Disease of Great Complexity*
Conduits, valved or nonvalved
Cyanotic congenital heart (all forms)
Double-outlet ventricle
Eisenmenger syndrome
Fontan procedure
Mitral atresia
Single ventricle (also called *double inlet* or *outlet*, *common* or *primitive*)
Pulmonary atresia (all forms)
Pulmonary vascular obstructive diseases
Transposition of the great arteries
Tricuspid atresia
Truncus arteriosus/hemitruncus
Other abnormalities of atrioventricular or ventriculoarterial connection not included above (i.e., crisscross heart, isomerism, heterotaxy syndromes, ventricular inversion)

From Webb G, Williams R, Alpert J, et al: 32nd Bethesda Conference: Care of the Adult with Congenital Heart Disease, October 2-3, 2000. J Am Coll Cardiol 37:1161-1198, 2001.

*These patients should be seen regularly at adult congenital heart disease centers.

TABLE 56–4 Cardiac Defects Causing Central Cyanosis	
Transposition of the great arteries	Ebstein's anomaly
Tetralogy of Fallot	Eisenmenger physiology
Tricuspid atresia	Critical pulmonary stenosis or atresia
Truncus arteriosus	Functionally single ventricle
Total anomalous pulmonary venous return	

Note 5 Ts and 2 Es.

The
End

Spectrum of Ventriculo-arterial Abnormality

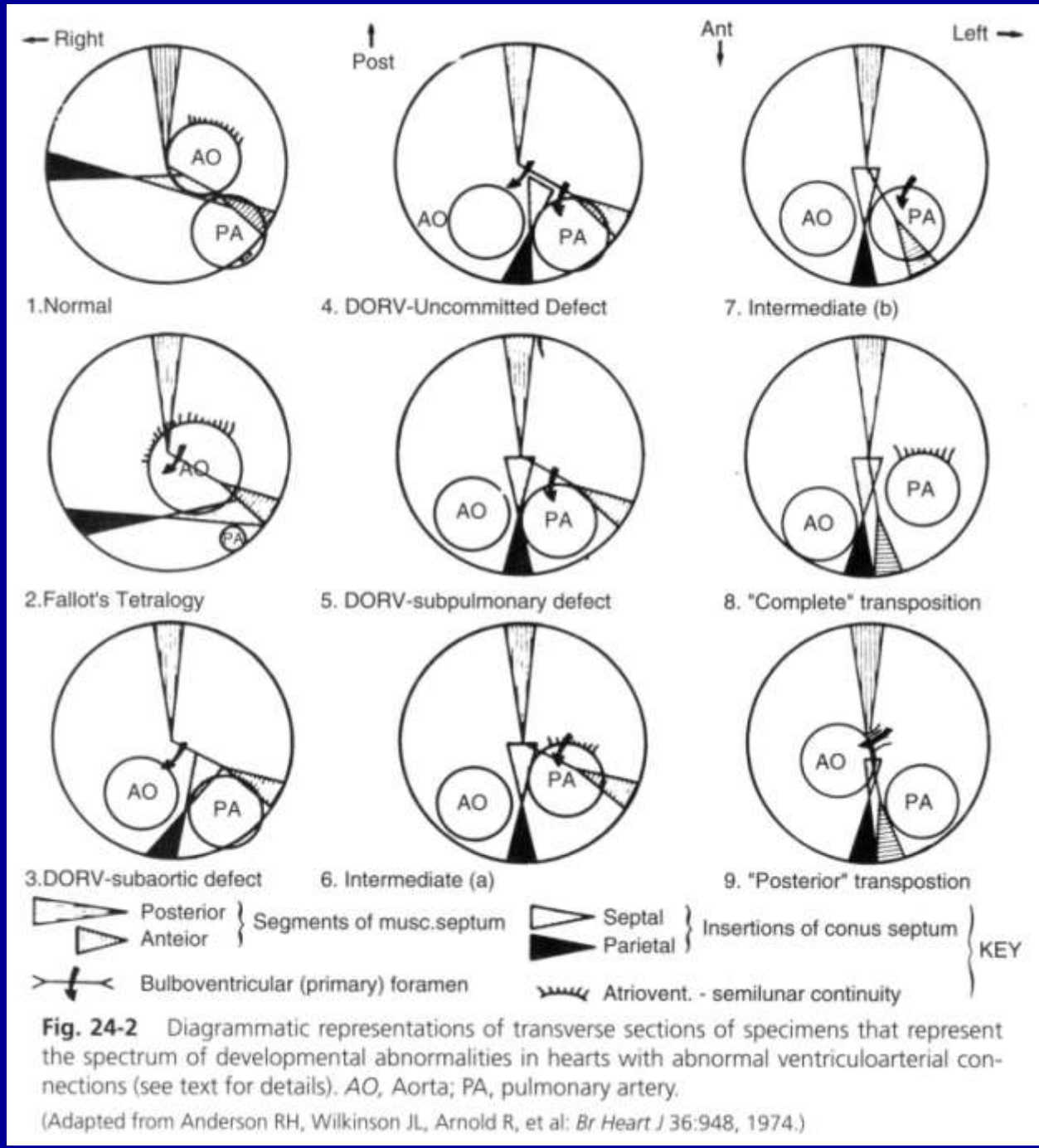


Fig. 24-2 Diagrammatic representations of transverse sections of specimens that represent the spectrum of developmental abnormalities in hearts with abnormal ventriculoarterial connections (see text for details). *AO,* Aorta; PA, pulmonary artery.

(Adapted from Anderson RH, Wilkinson JL, Arnold R, et al: *Br Heart J* 36:948, 1974.)

Mavroudis, 2003, p. 412

Topics in Congenital Heart Disease

- Genetic abnormalities causing cardiovascular disease
- **Pathology, pathophysiology, recognition, and treatment**
- Congenital heart disease in adults

5 Basic Questions In Congenital Heart Disease

- Is the patient acyanotic or cyanotic?
- Is pulmonary arterial blood flow increased or not?
- Does the malformation originate in the left or right side of the heart?
- Which is the dominant ventricle?
- Is pulmonary hypertension present or not?

Perloff, 1994, p. 7

Genetic Abnormalities

- Many genes are being identified that affect laterality

Congenital Heart Disease Classification - 1

- Intracardiac systemic-pulmonary communications (acyanotic)
- Extracardiac systemic-pulmonary communications (acyanotic)
- Left heart valve/vessel malformations
- Right heart valve/vessel malformations
- Pulmonary venous connection

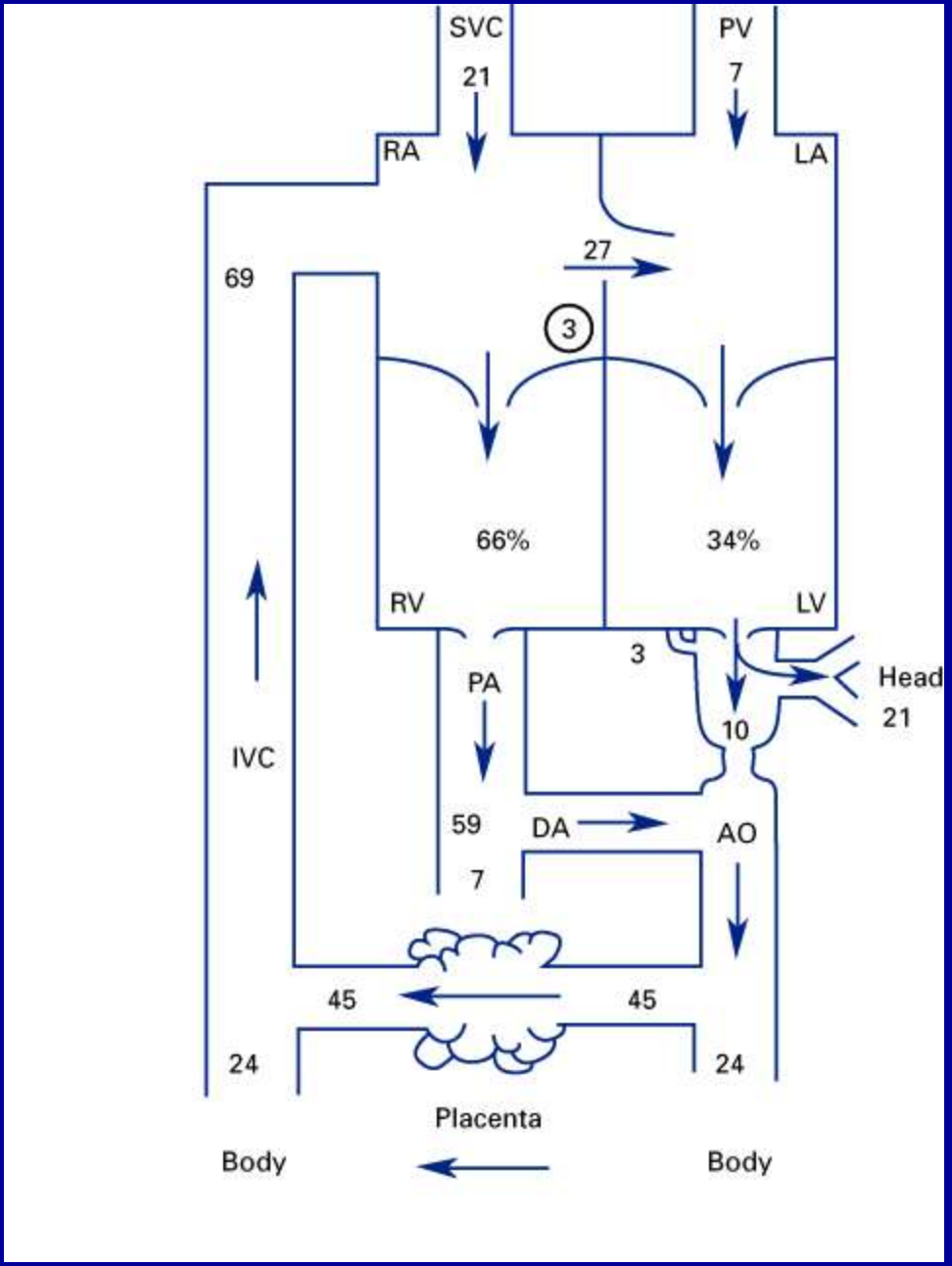
Congenital Heart Disease Classification - 2

- Cardiac malpositions
- Coronary malformations

Fetal Circulation near term

Numbers represent percent of combined ventricular output

FLOW

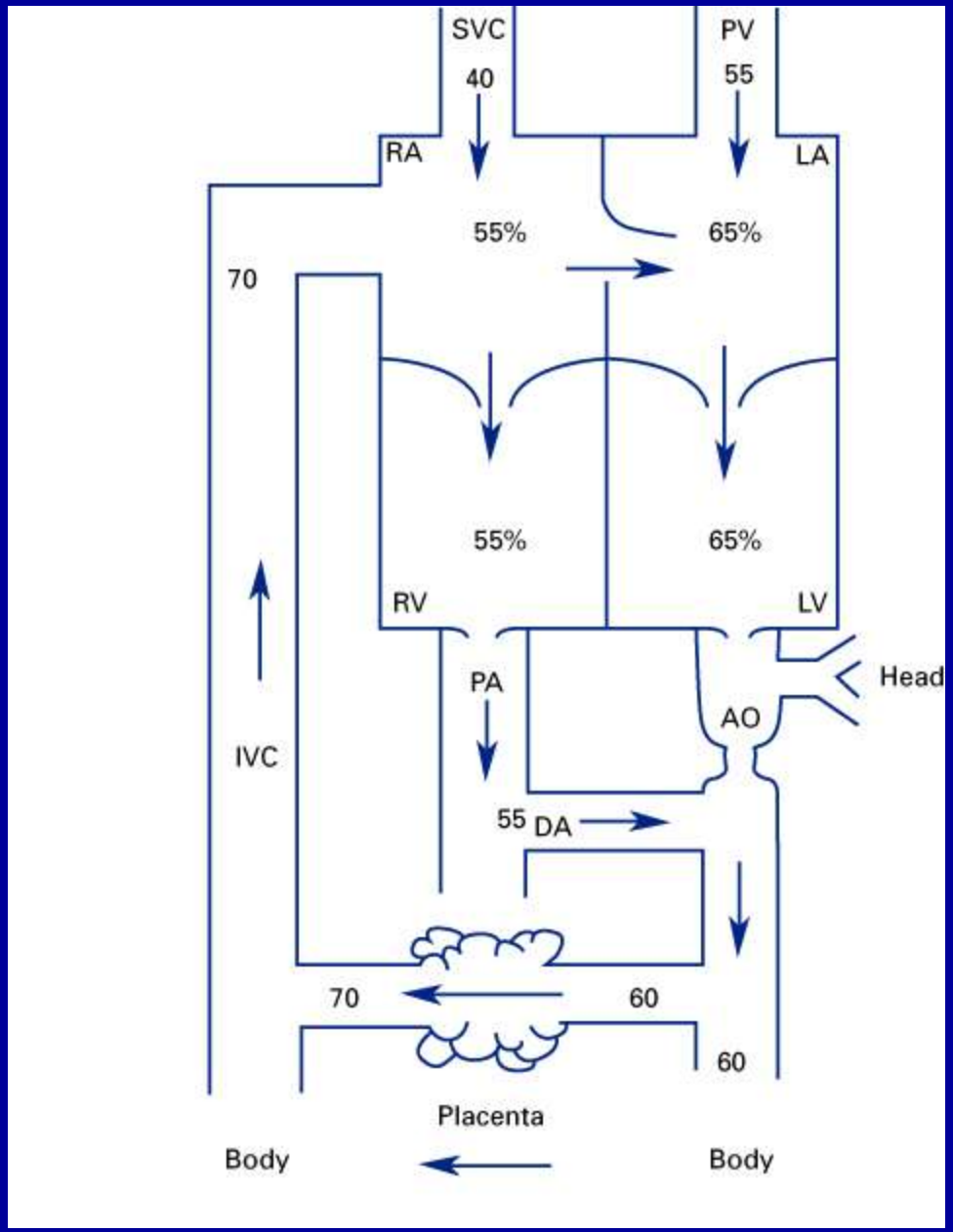


From Hurst, 1999, Ch 70

Fetal Circulation near term

Numbers represent oxygen saturation

SATURATION

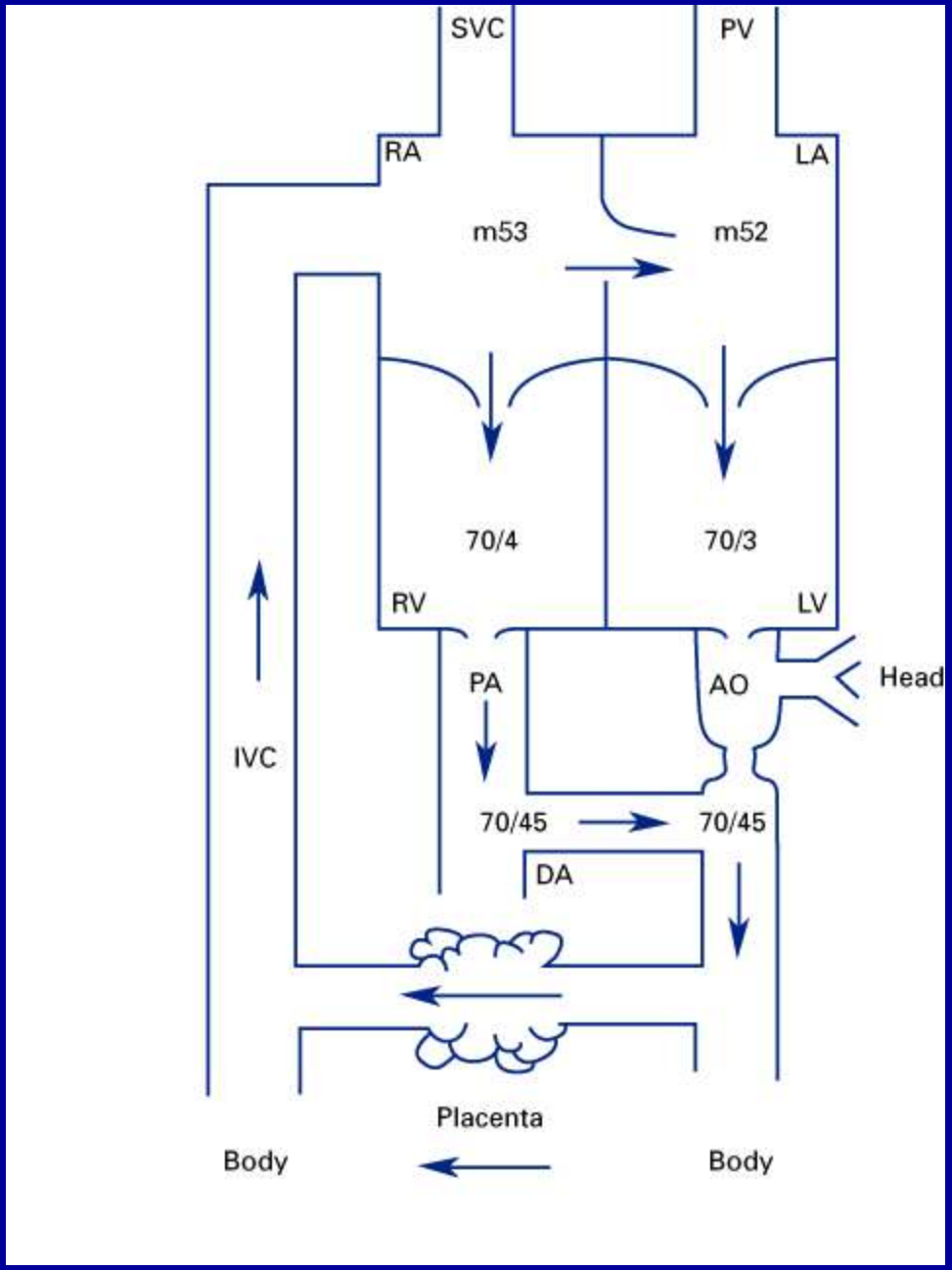


From Hurst, 1999, Ch 70

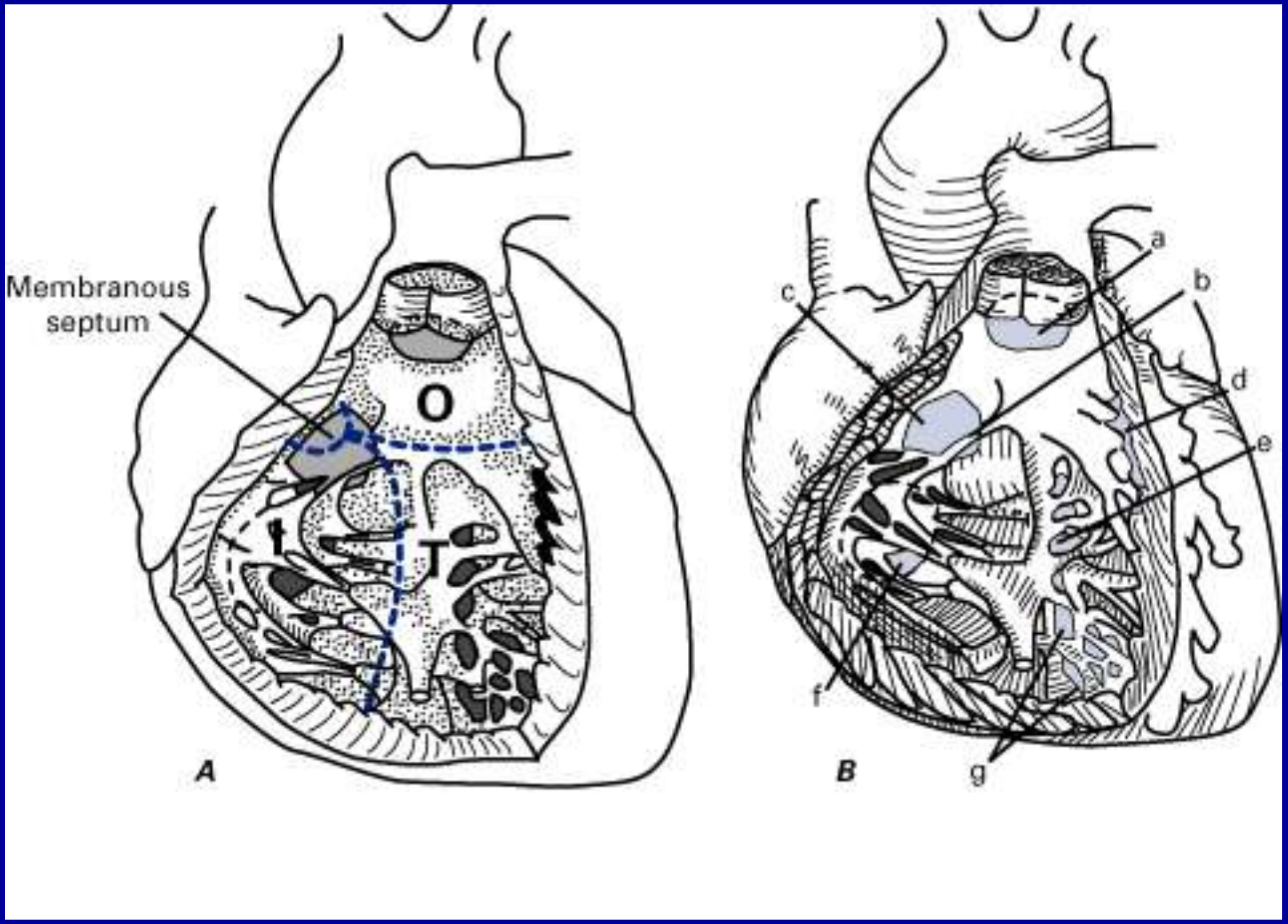
Fetal Circulation near term

Numbers represent cardiovascular pressures

PRESSURE

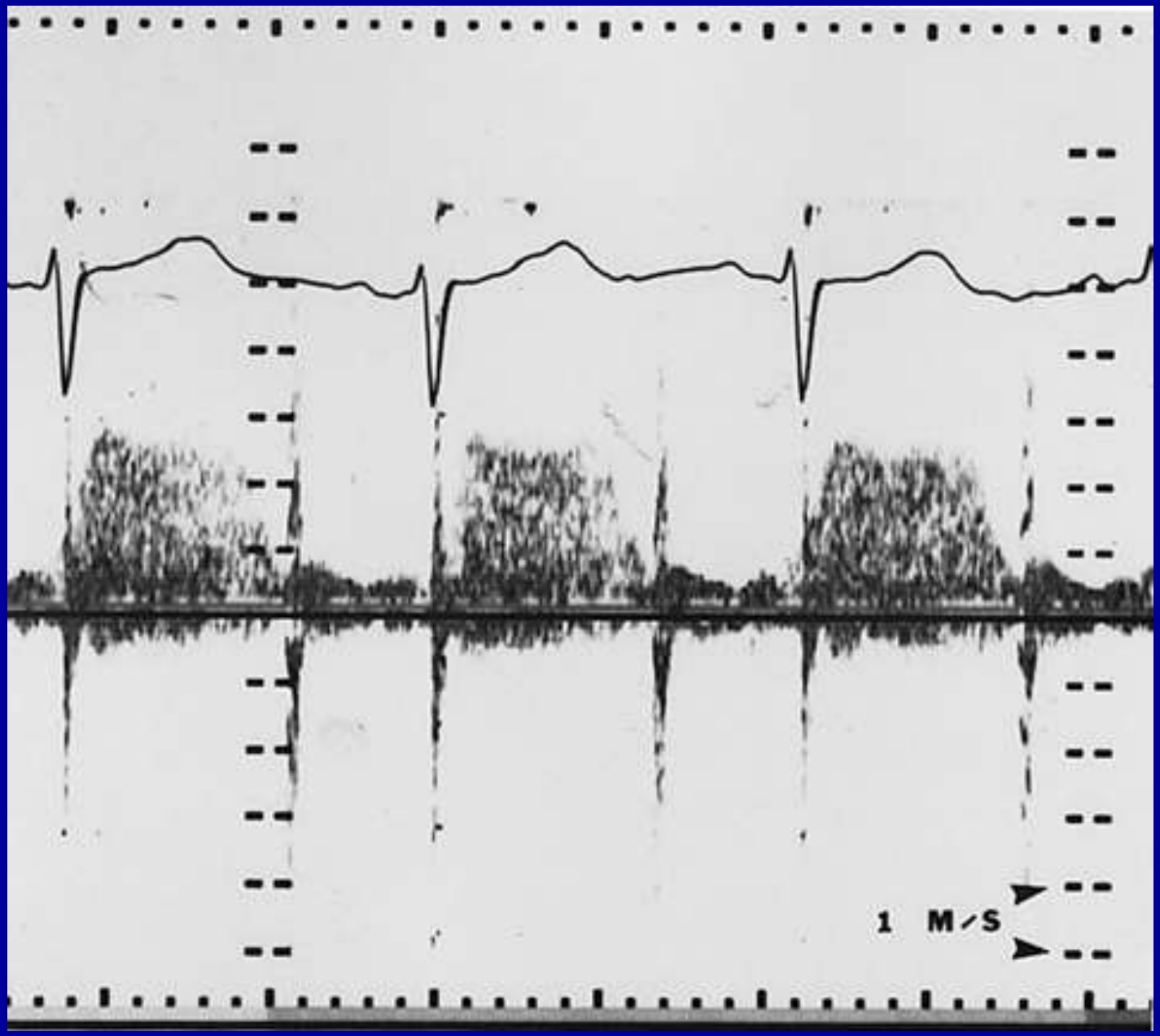


From Hurst, 1999, Ch 70



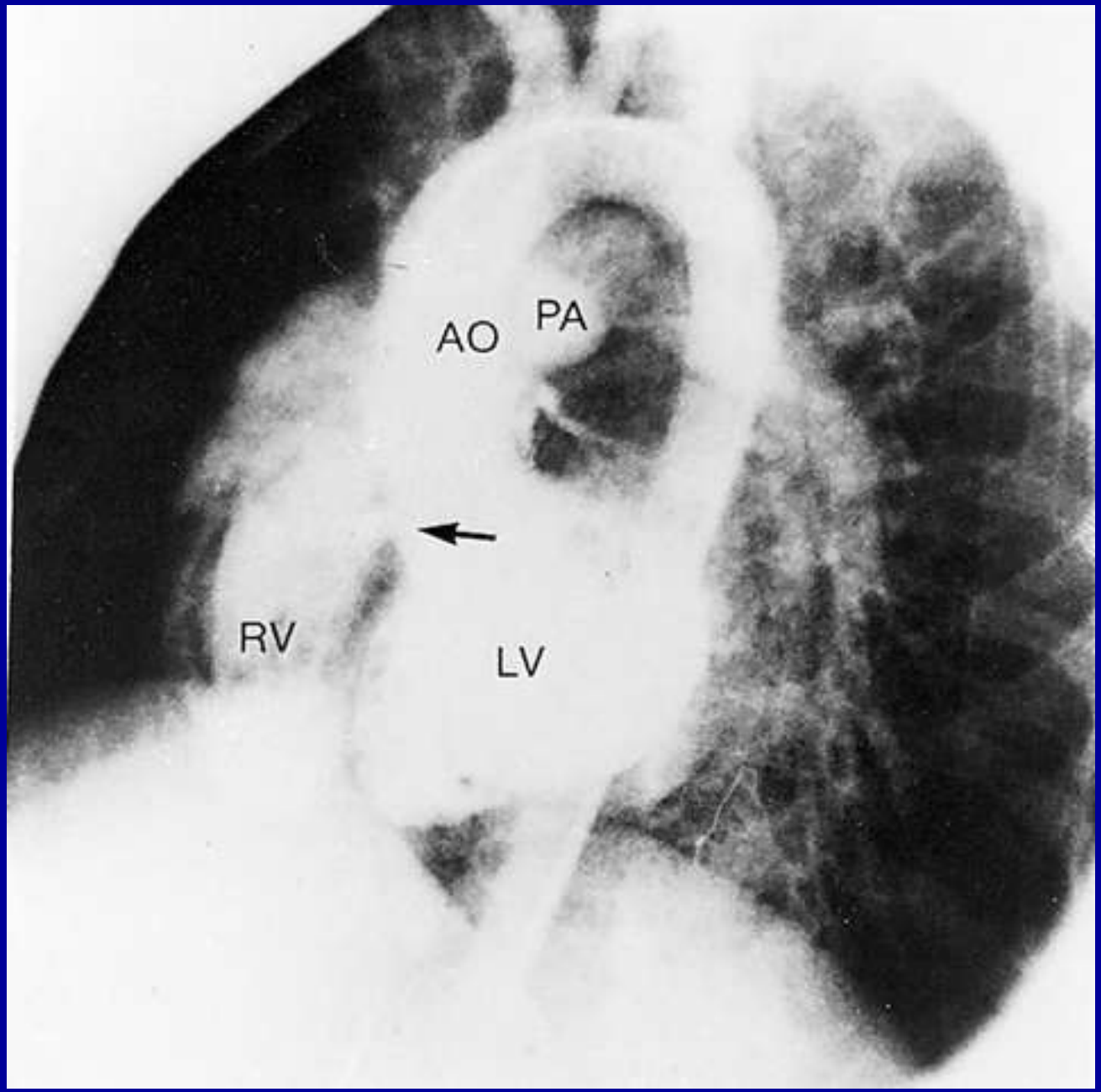
From Hurst, 1999, Ch 70

CW Doppler, VSD



From Hurst, 1999, Ch 70

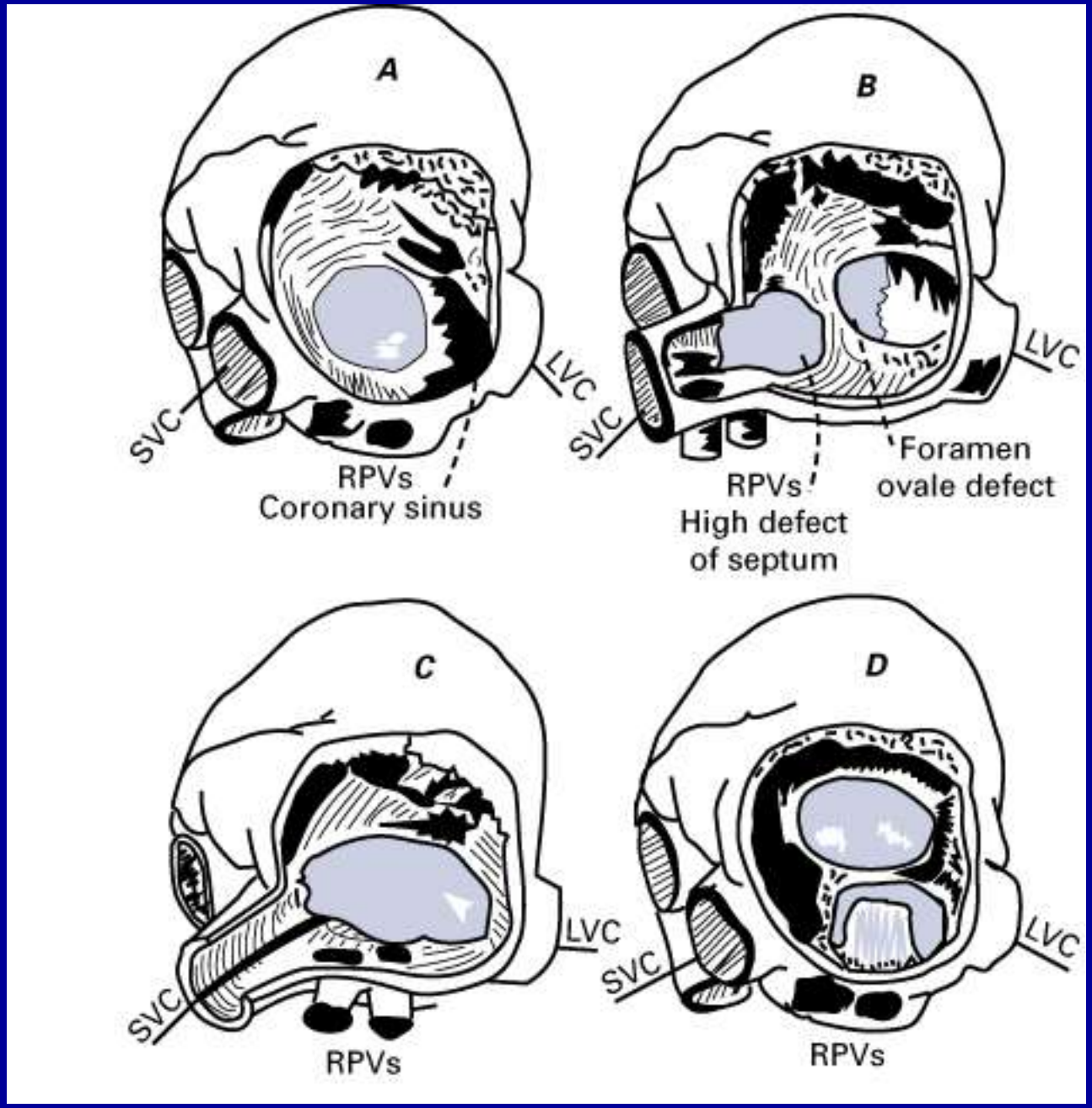
LV angiogram
LAO projection
VSD



From Hurst, 1999, Ch 70

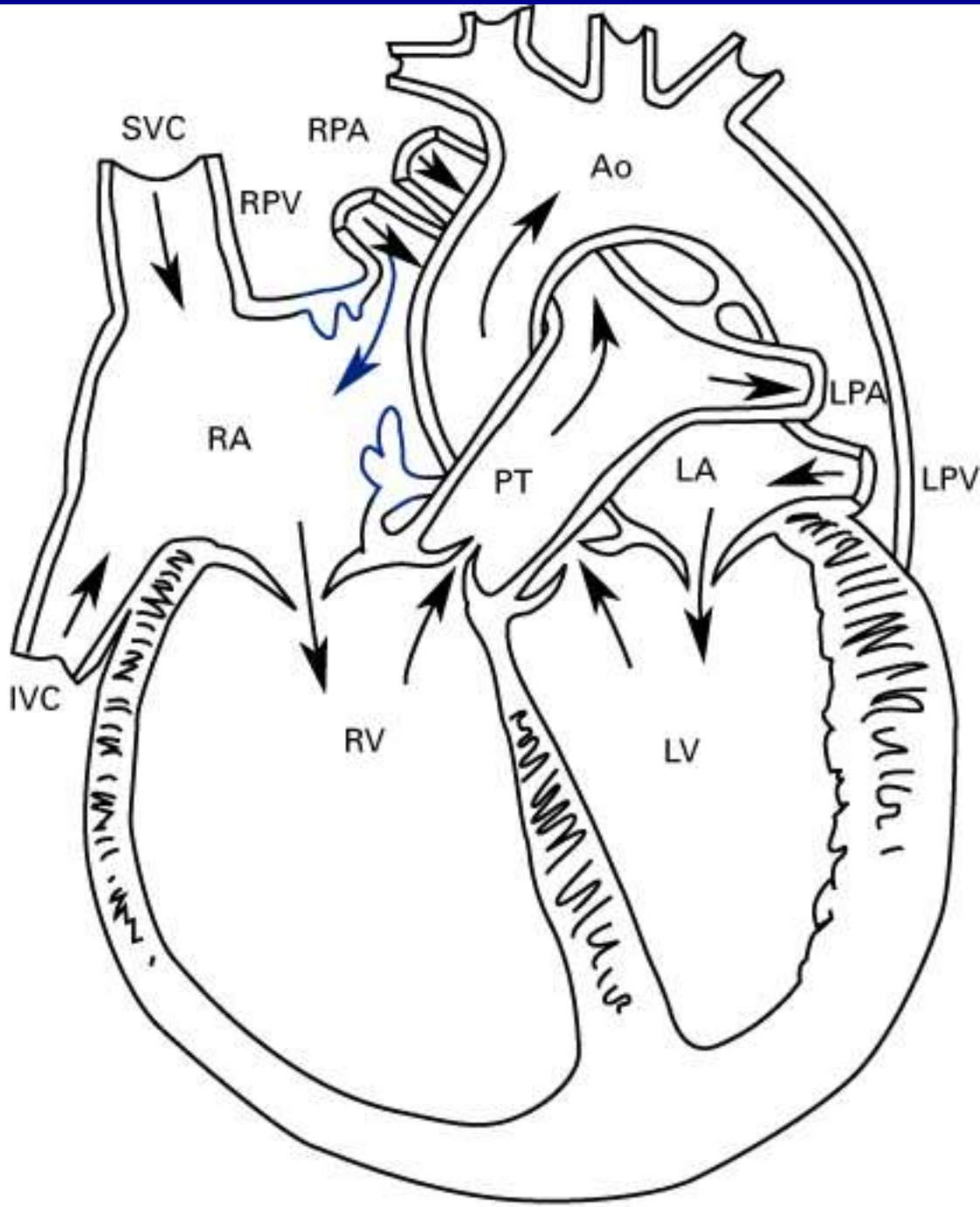
ASD types

A: secundum
B: sinus venosus
C: large
secundum
D: primum,
partial AV canal



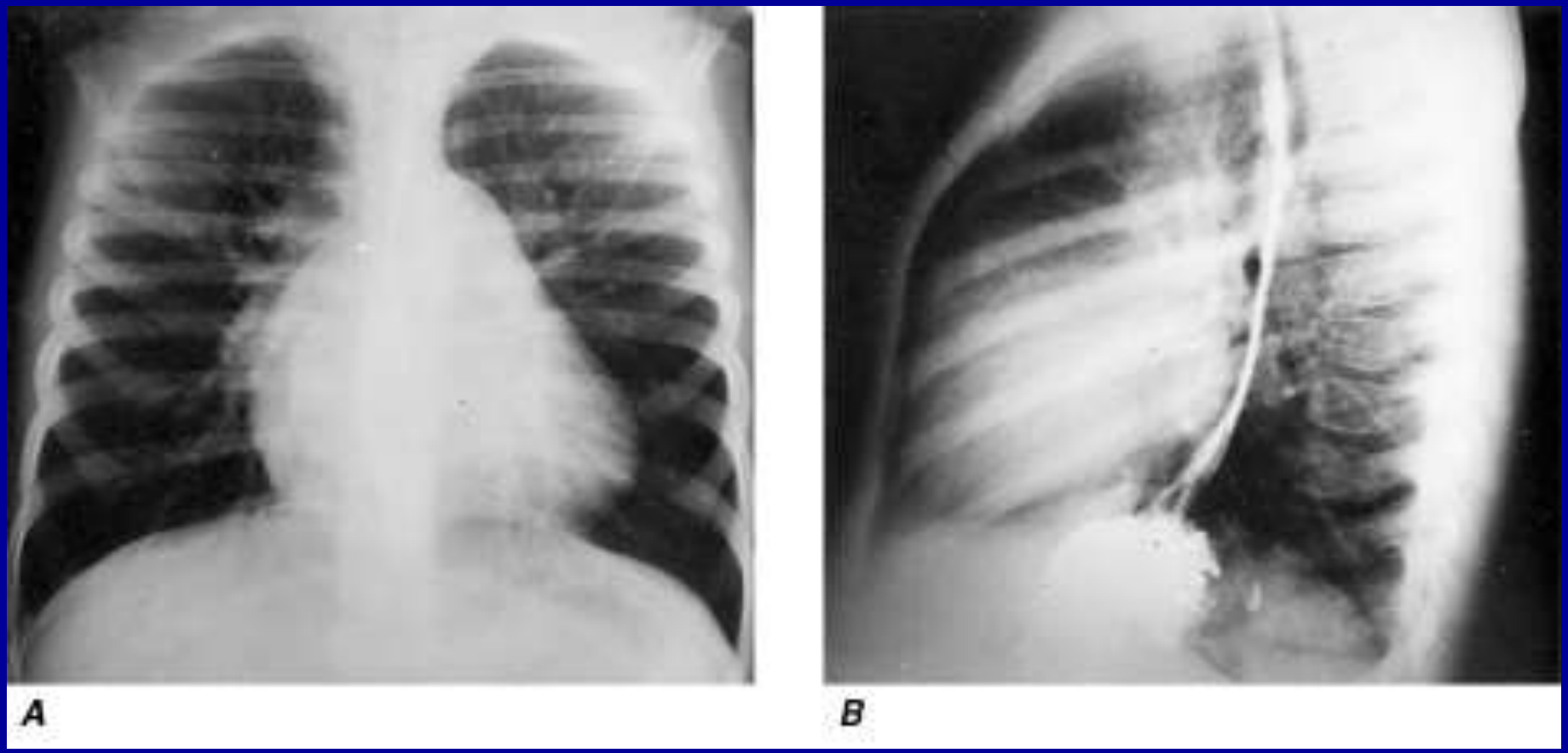
From Hurst, 1999, Ch 70

ASD at
fossa ovalis
(secundum)

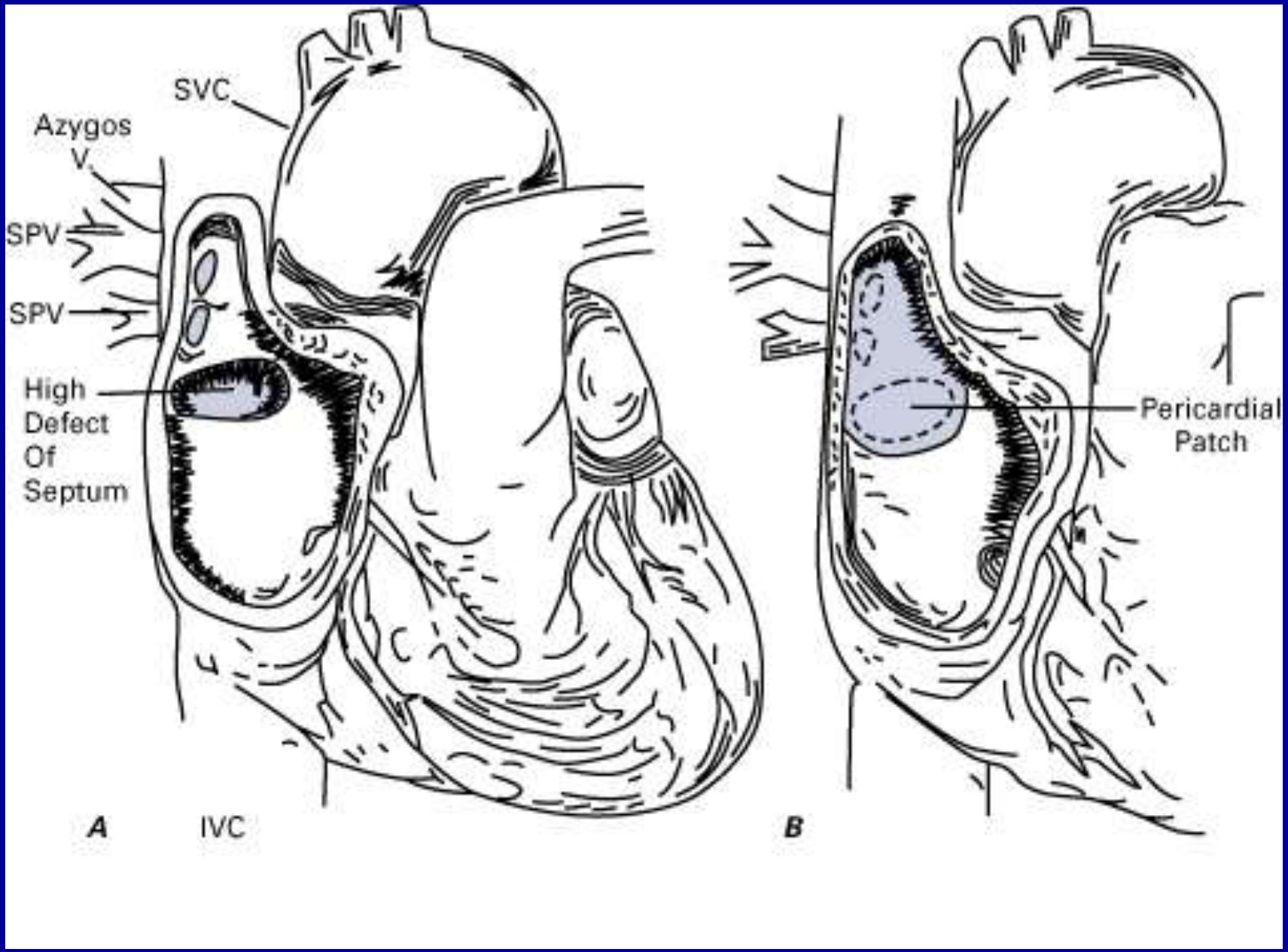


From Hurst, 1999
Ch 70

CXR in ASD with large L to R shunt and no pulm htn, 4 y.o.

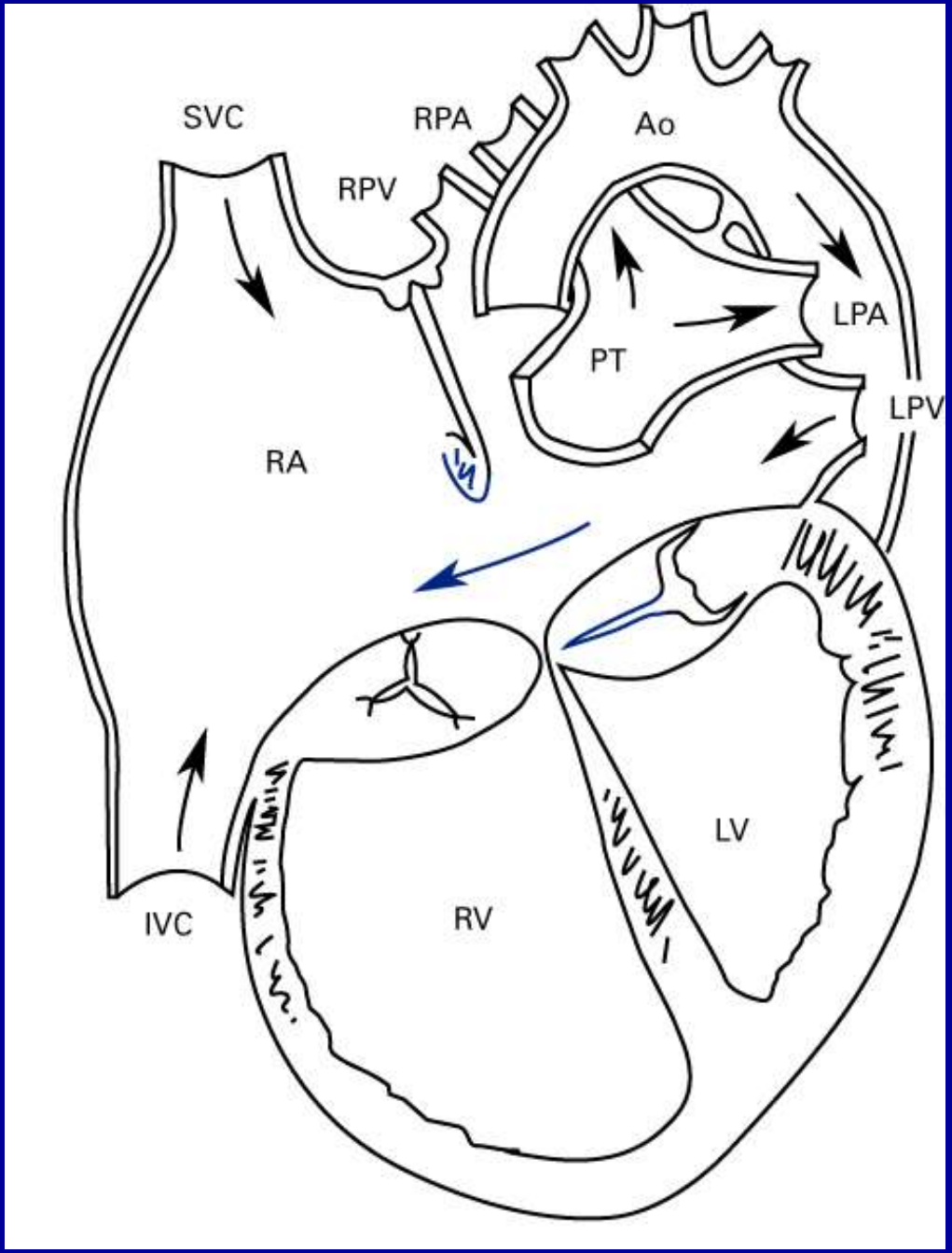


From Hurst, 1999, Ch 70



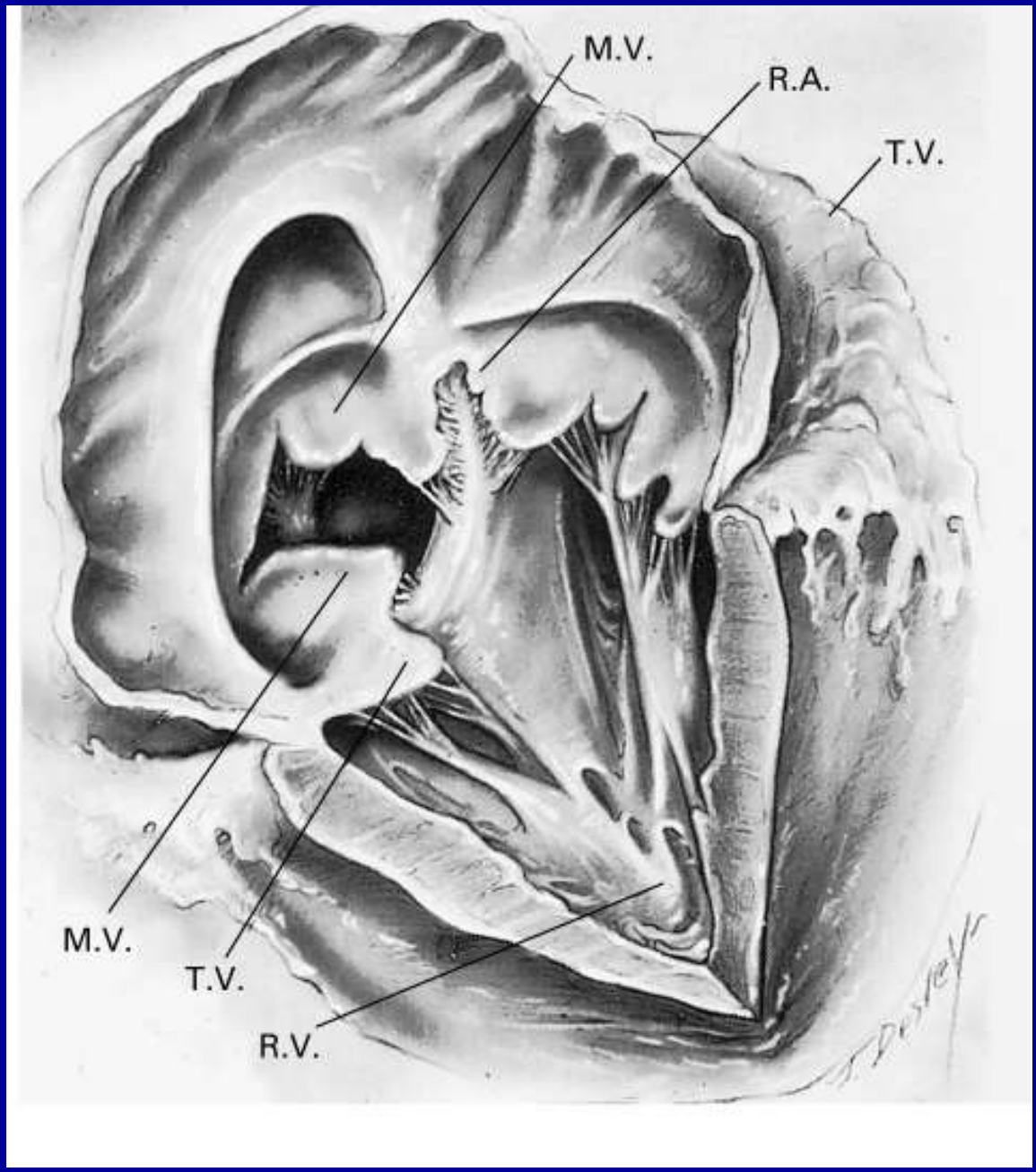
From Hurst, 1999, Ch 70, Sinus venosus ASD and its repair

Partial common AV canal
cleft anterior MV leaflet
normal TV leaflet



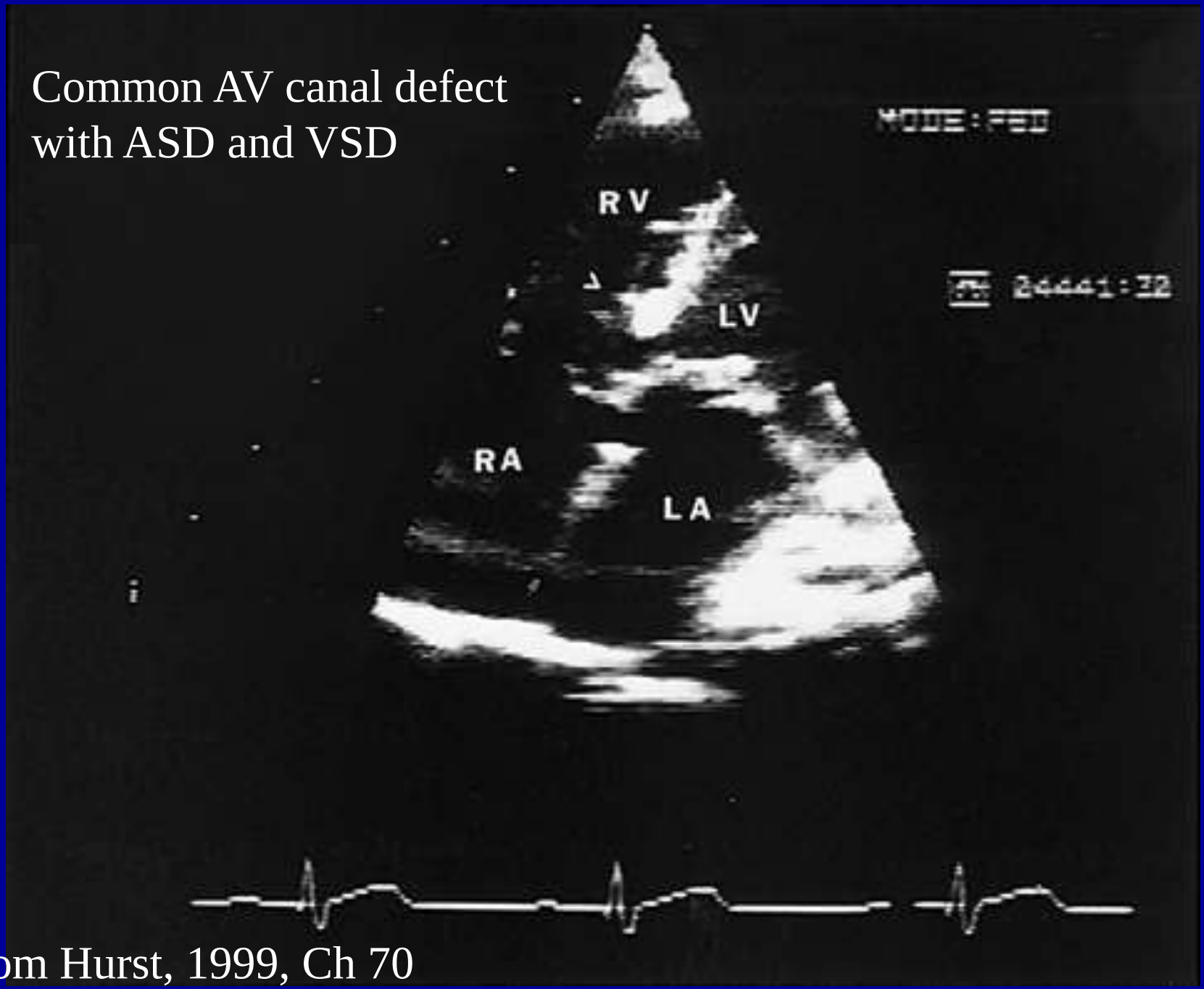
From Hurst, 1999, Ch 70

Complete Common AV Canal



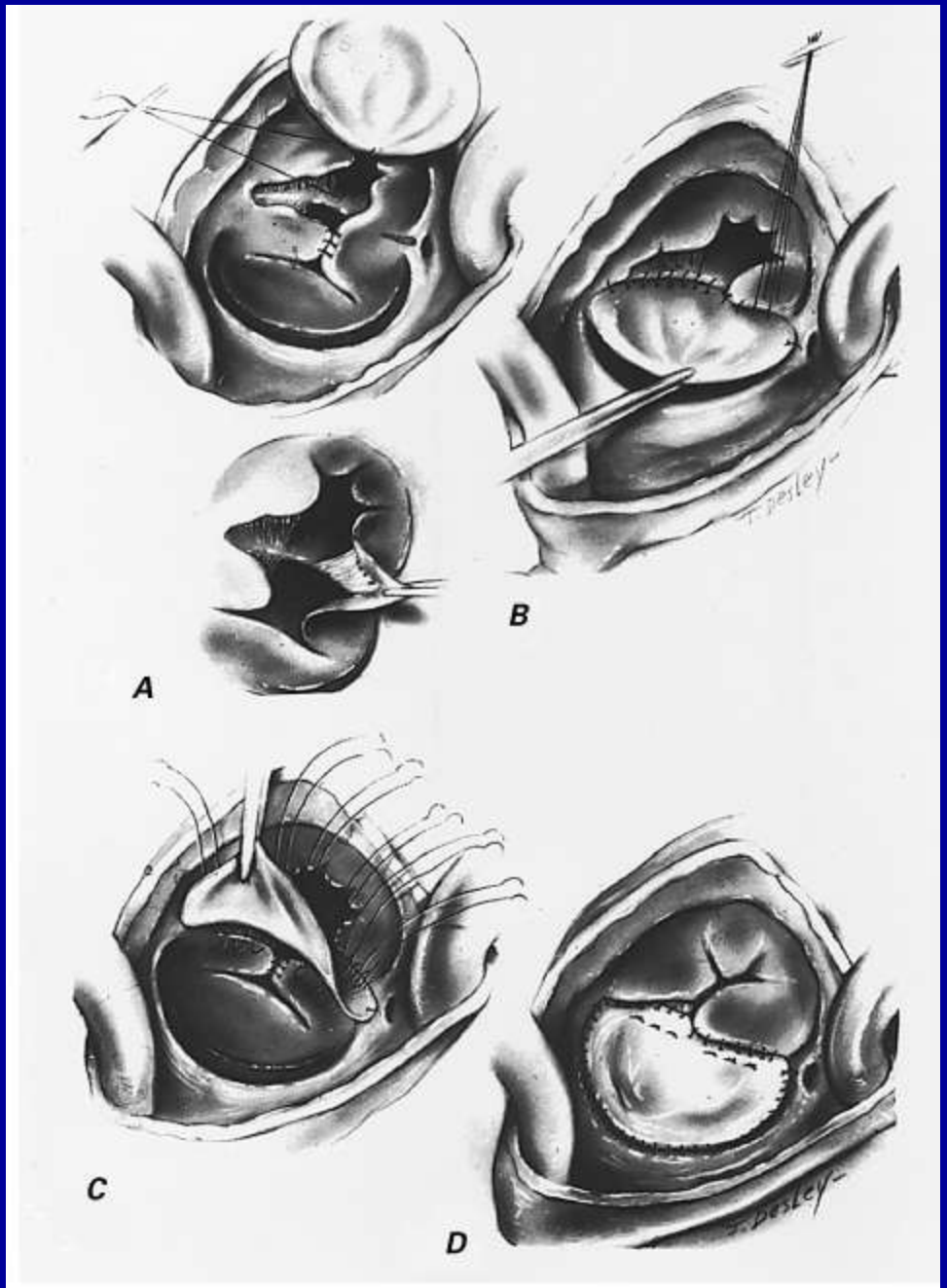
From Hurst, 1999, Ch 70

Common AV canal defect with ASD and VSD



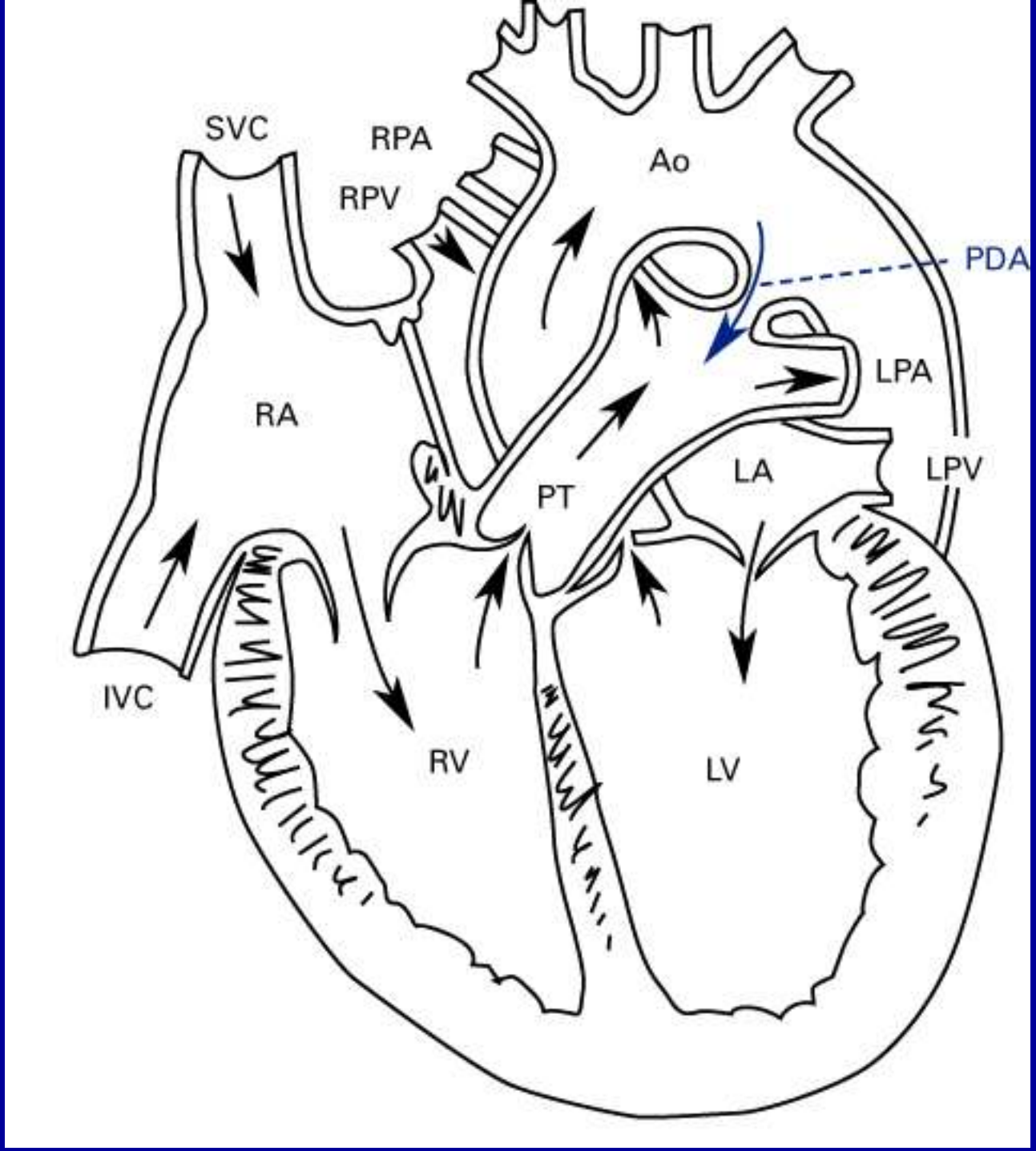
From Hurst, 1999, Ch 70

Surgery for common AV canal
pericardial patch

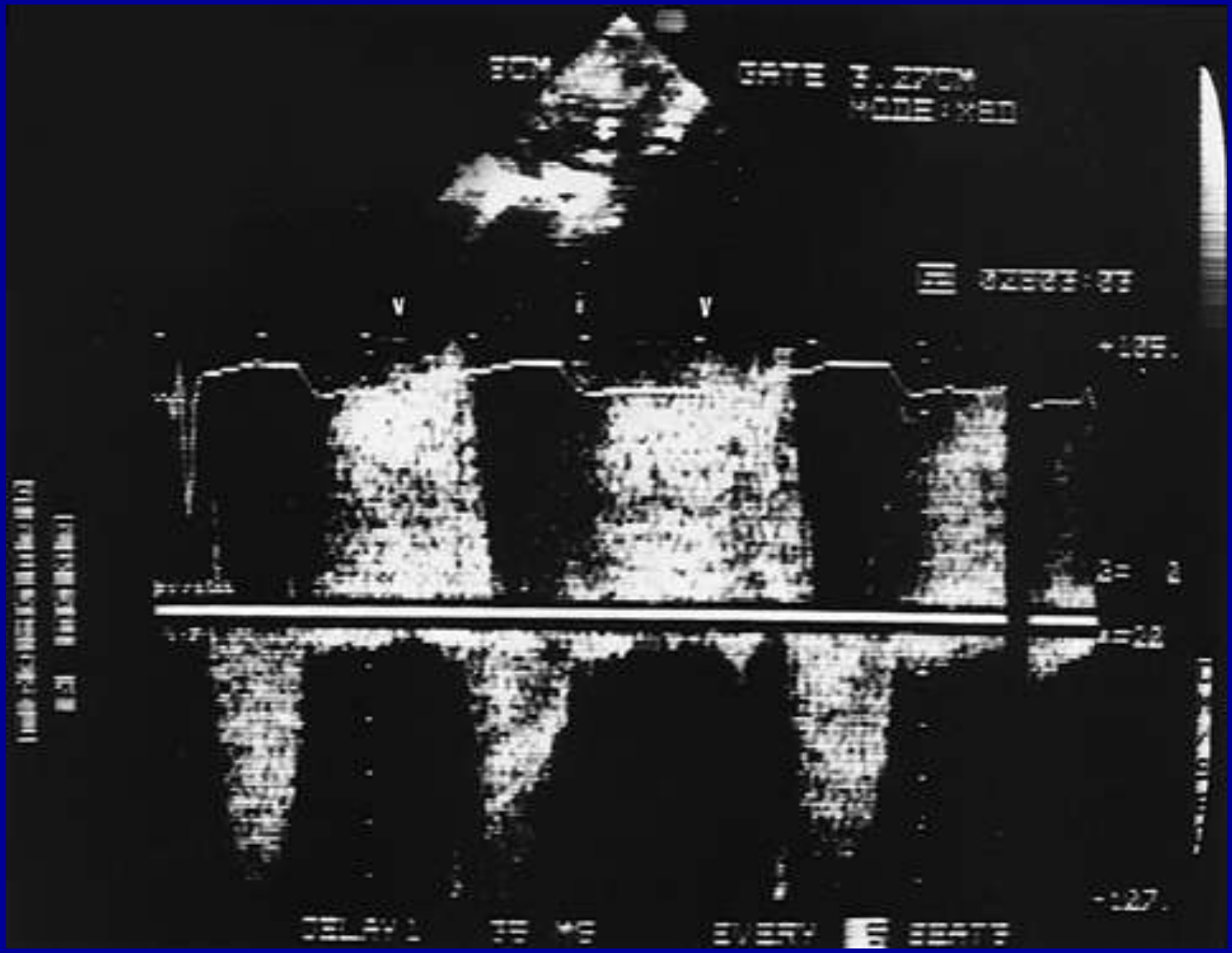


From Hurst, 1999, Ch 70

Patent ductus arteriosus

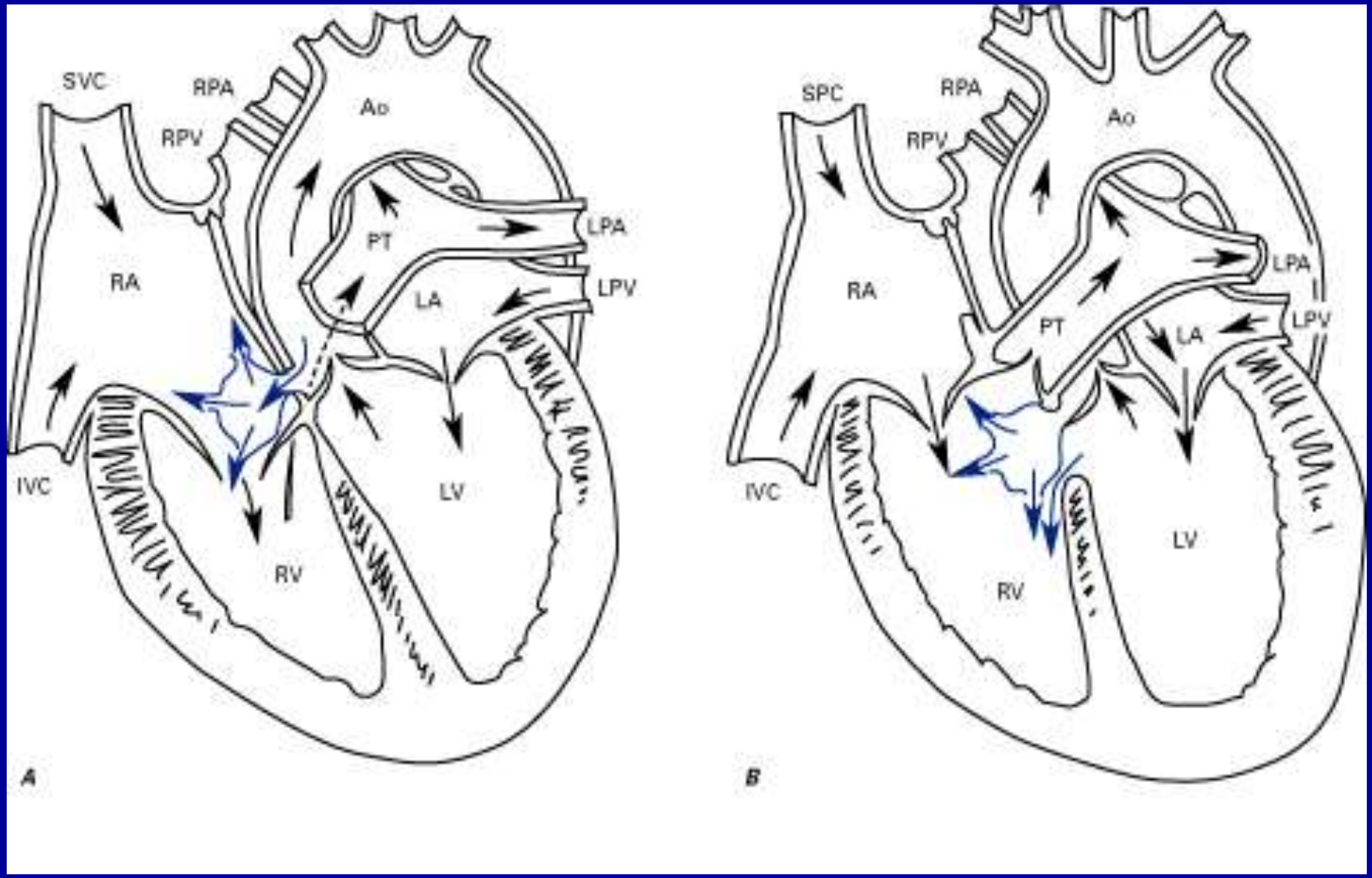


From Hurst, 1999
Ch 70



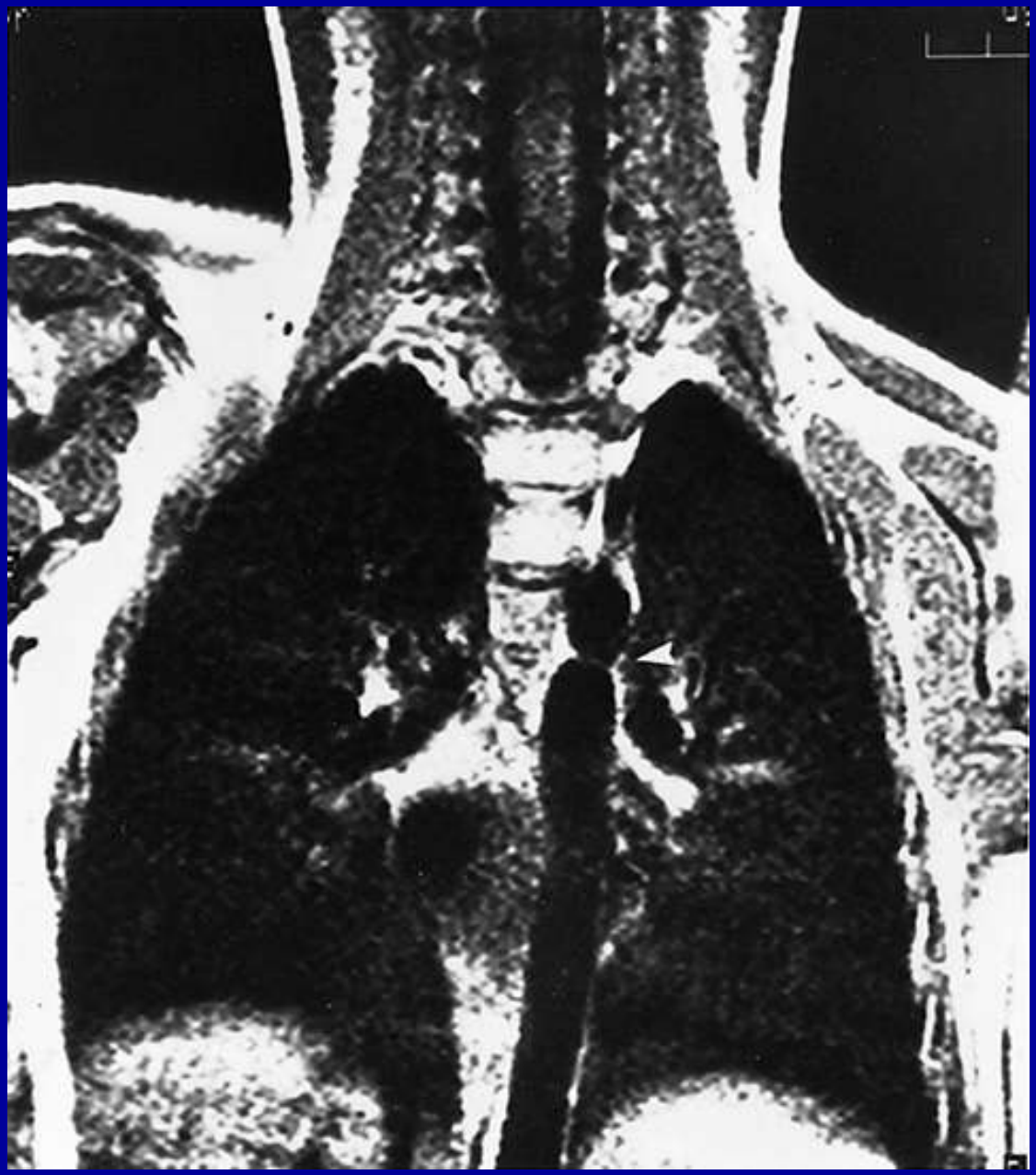
From Hurst, 1999, Ch 70 PDA flow in PW Doppler in Pulm Artery

Sinus of Valsalva fistula: A=posterior sinus to RA; B=right sinus to RV



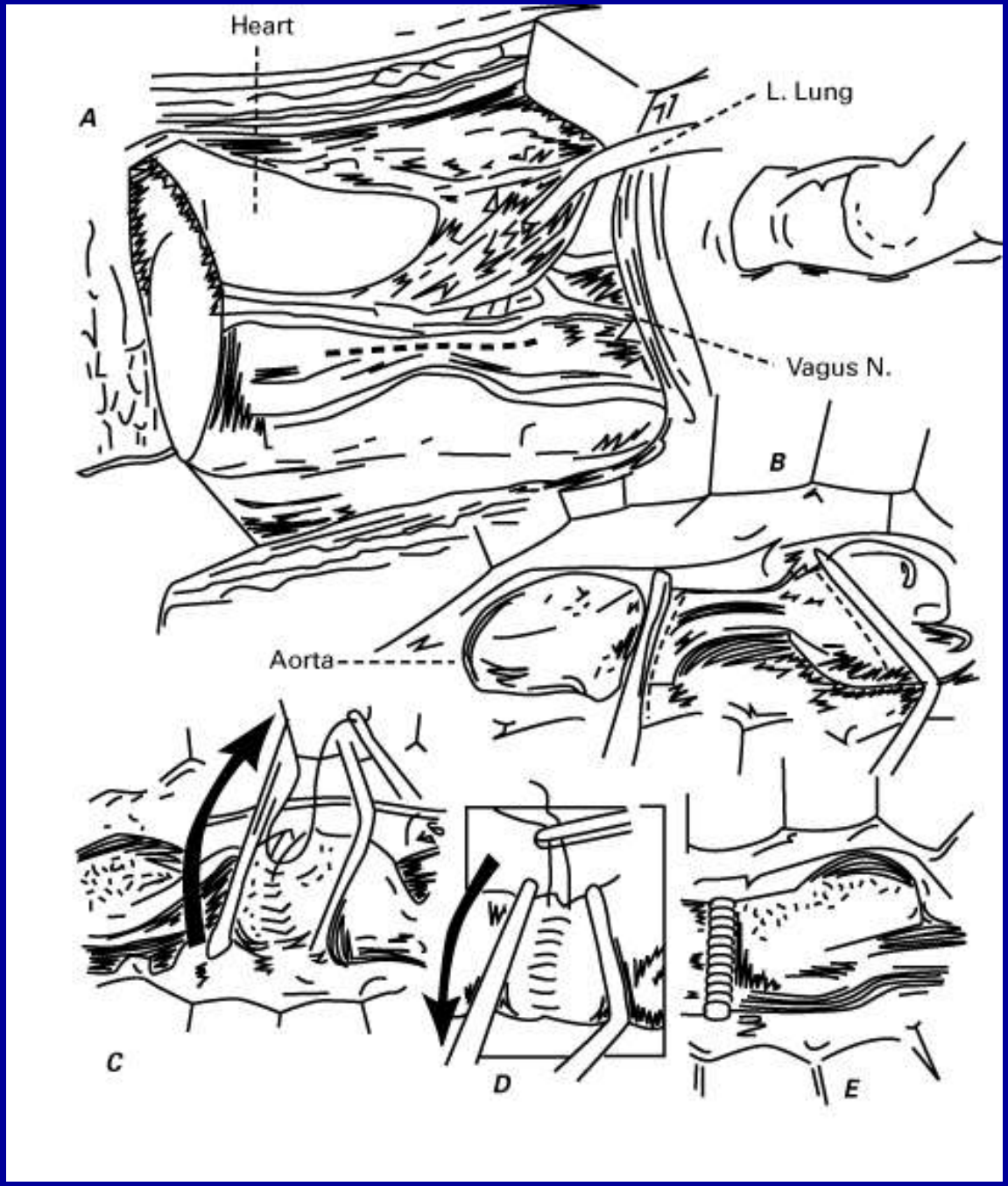
From Hurst, 1999, Ch 70

MRI of
Coarctation of Aorta



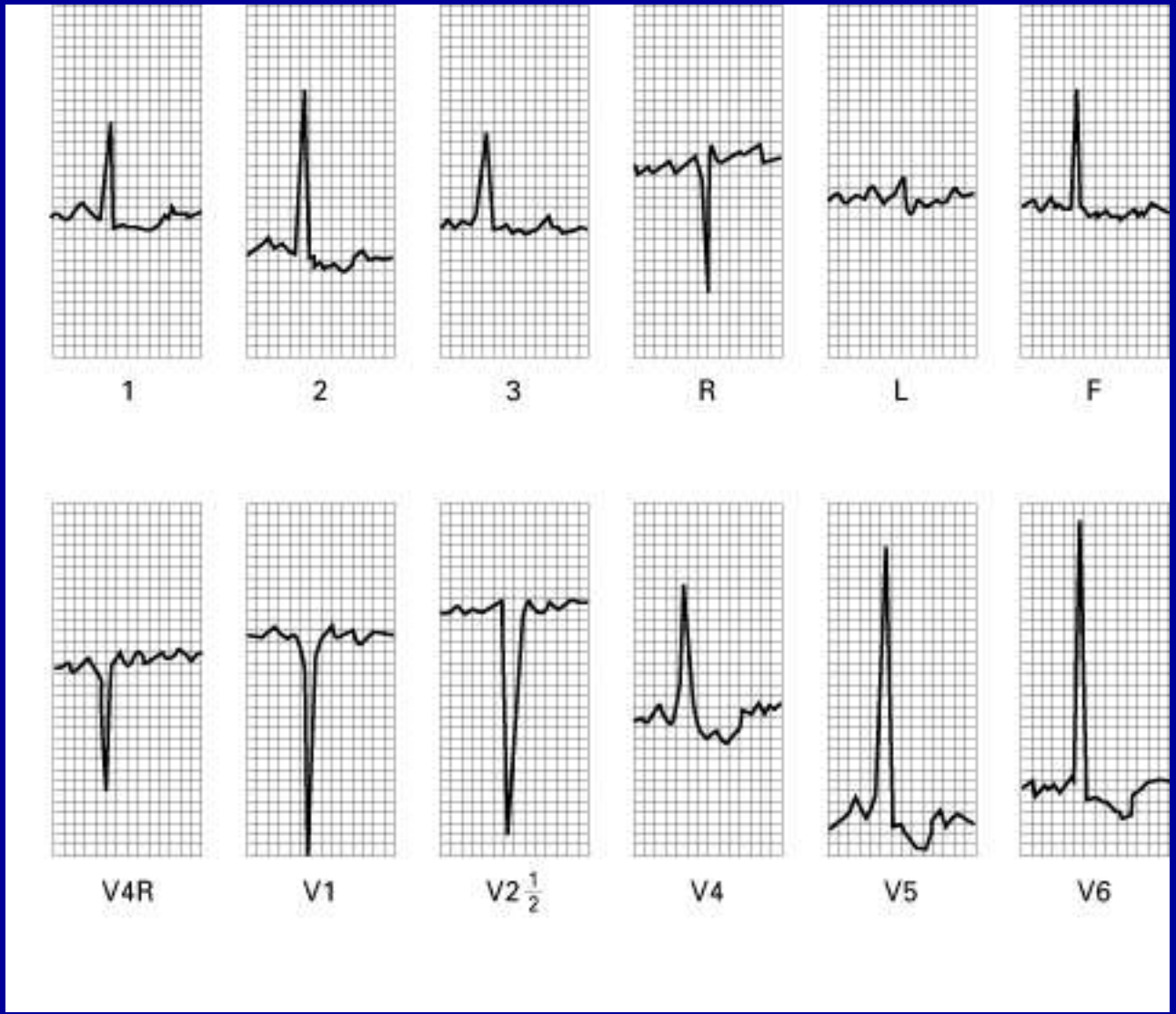
From Hurst, 1999, Ch 70

Repair of Coarctation at Surgery



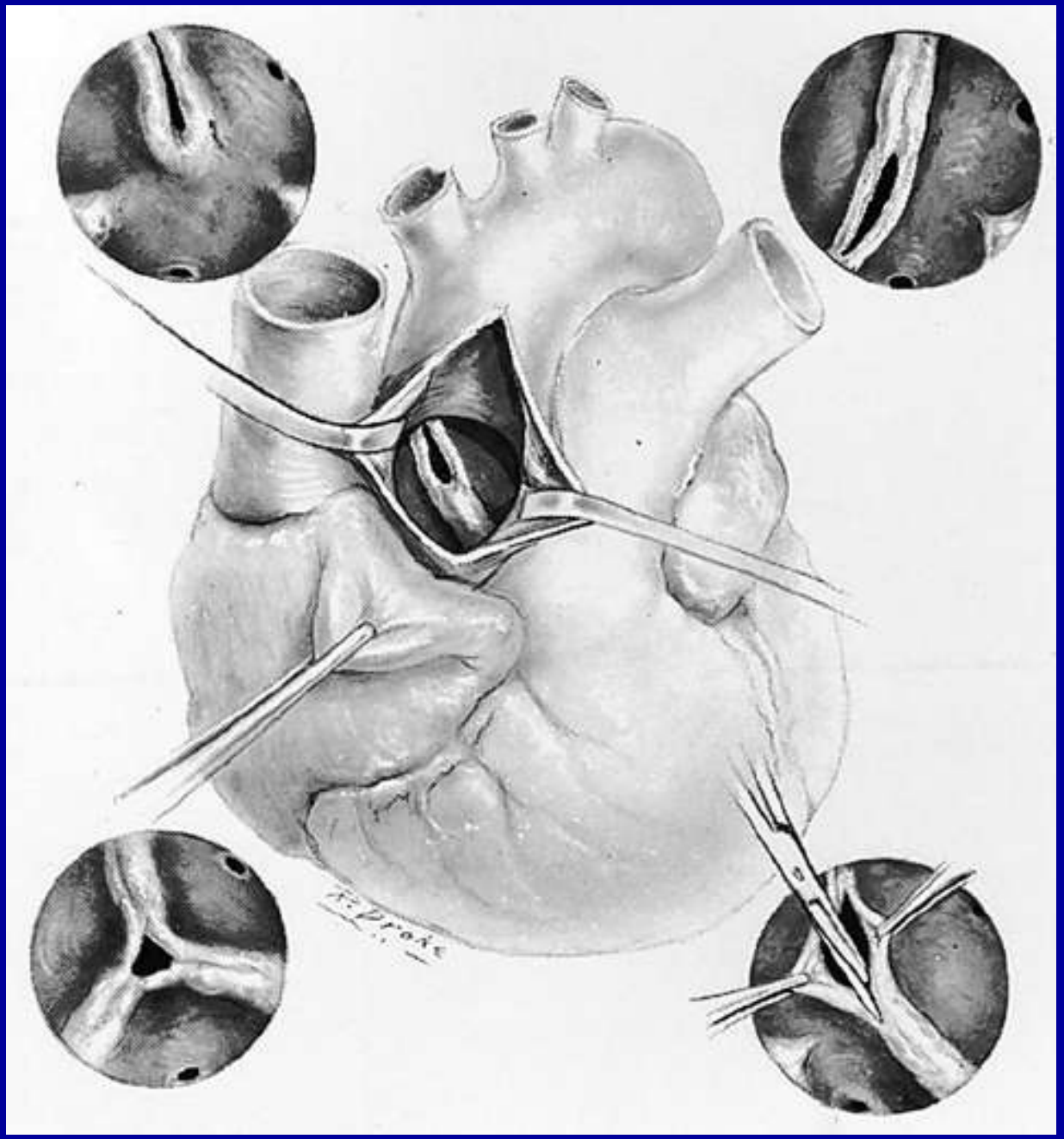
From Hurst, 1999, Ch 70

Congenital Valvular AS in an 8 y.o. boy



From Hurst, 1999, Ch 70

Valvular deformity types in Congenital Valvular Aortic Stenosis



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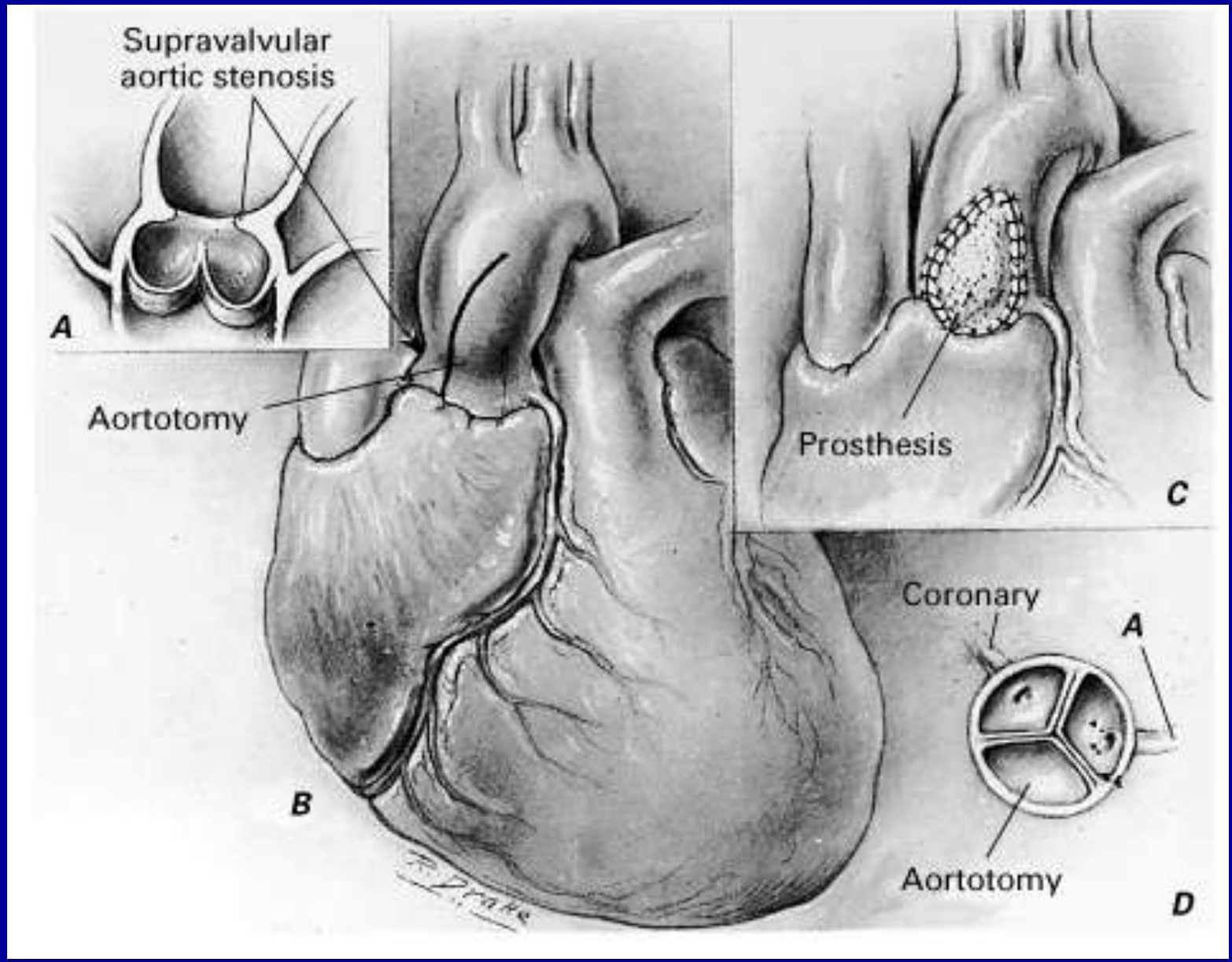

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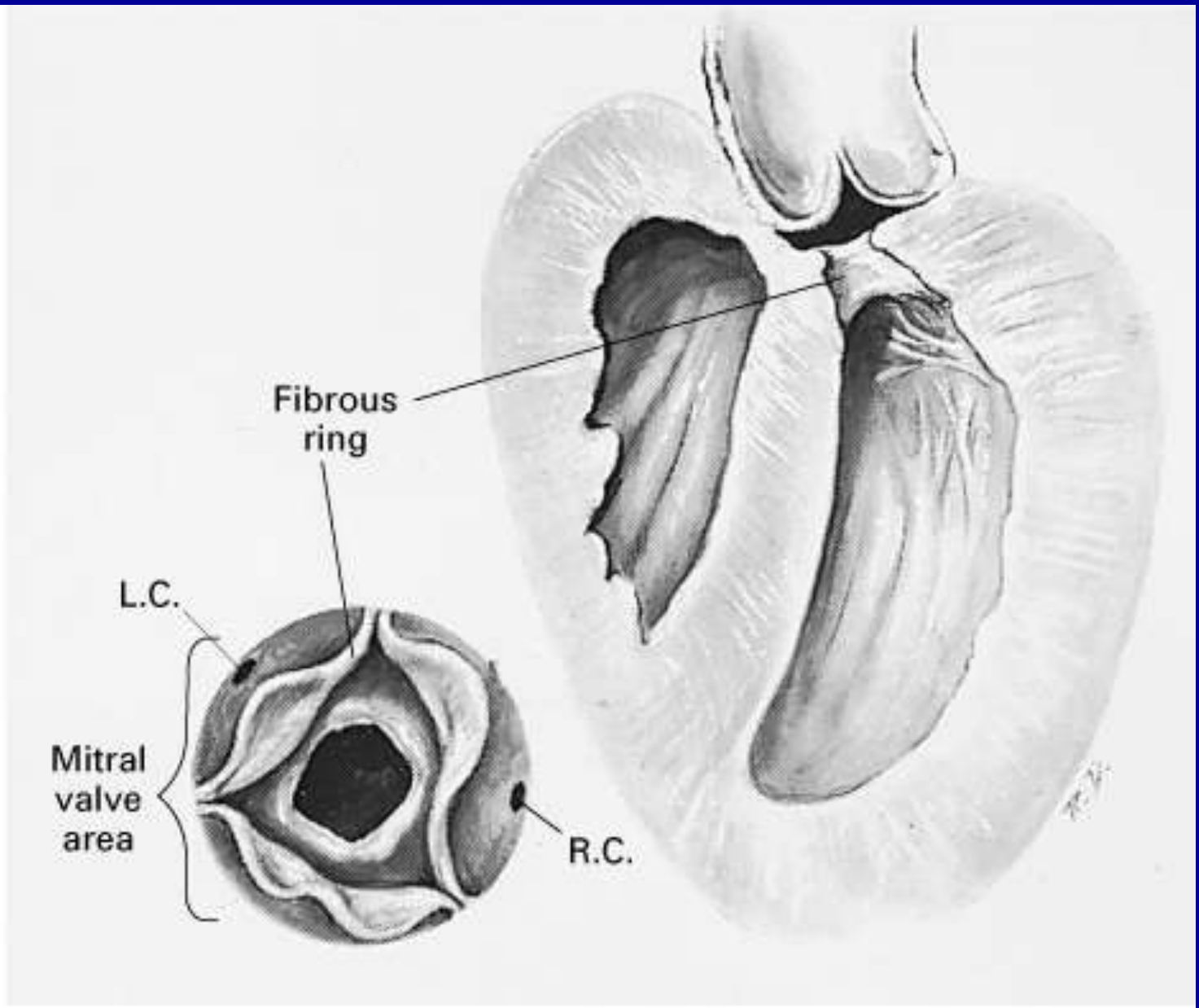
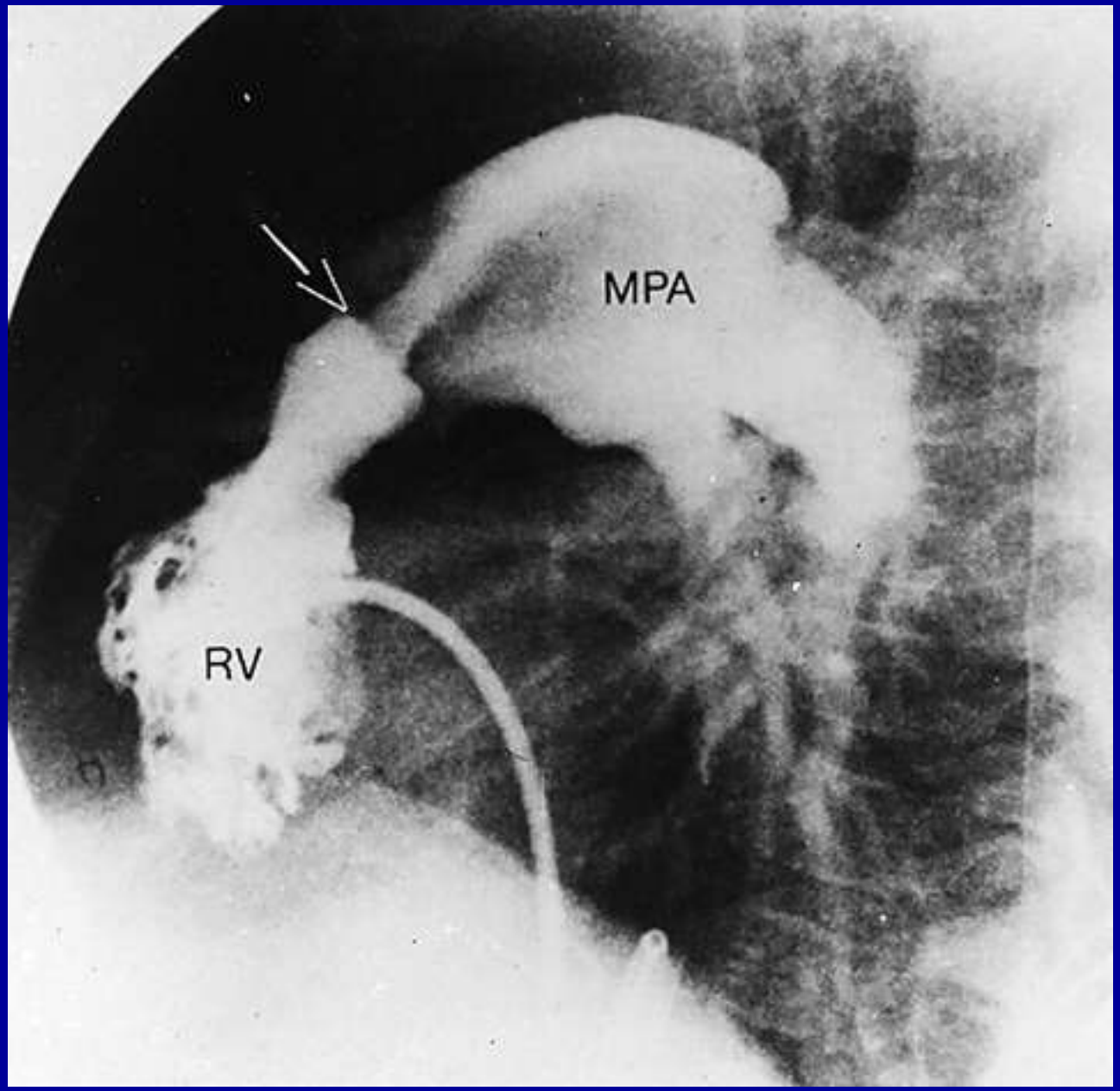


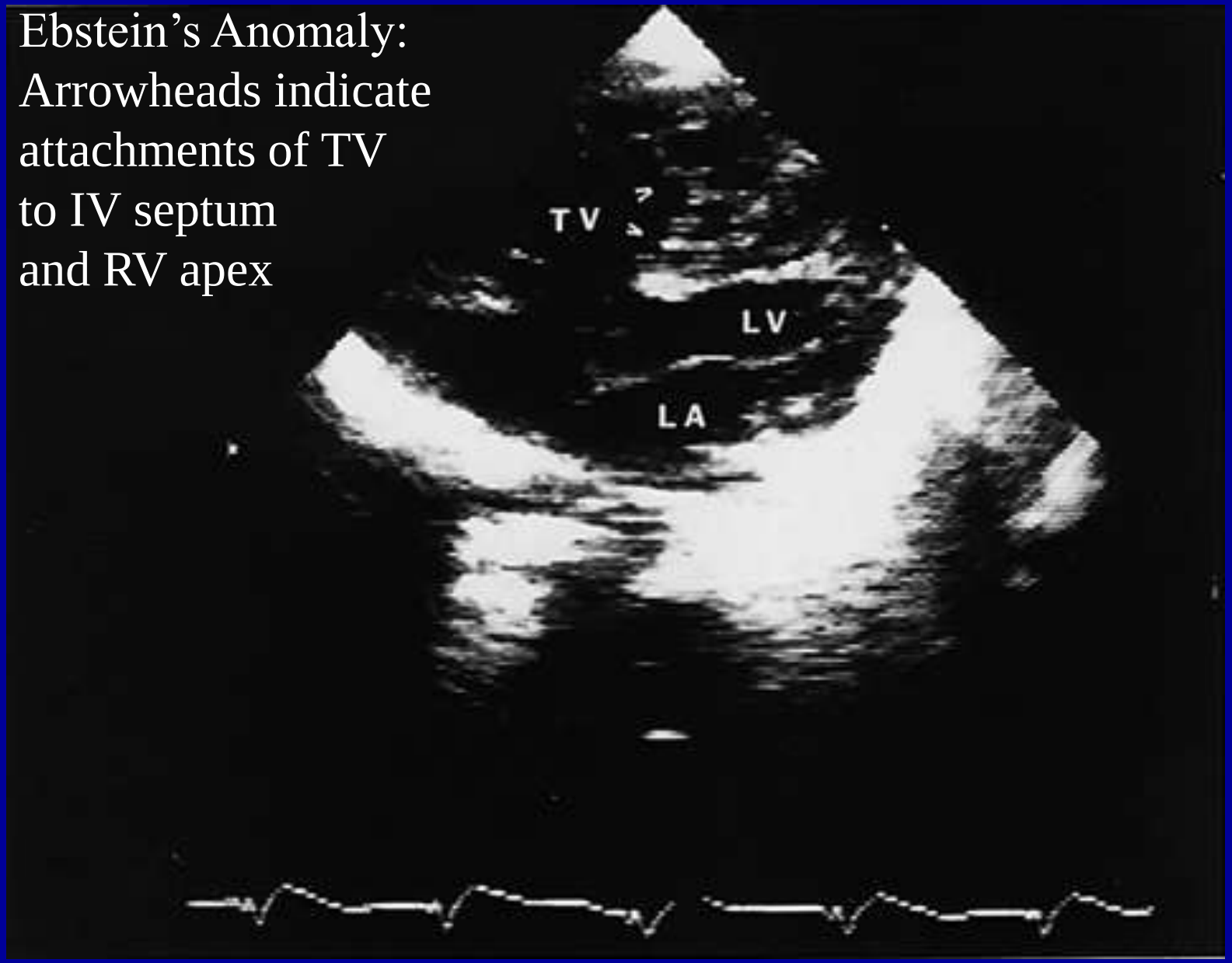
From Hurst, 1999, Ch 70

Valvular
Pulmonic
Stenosis

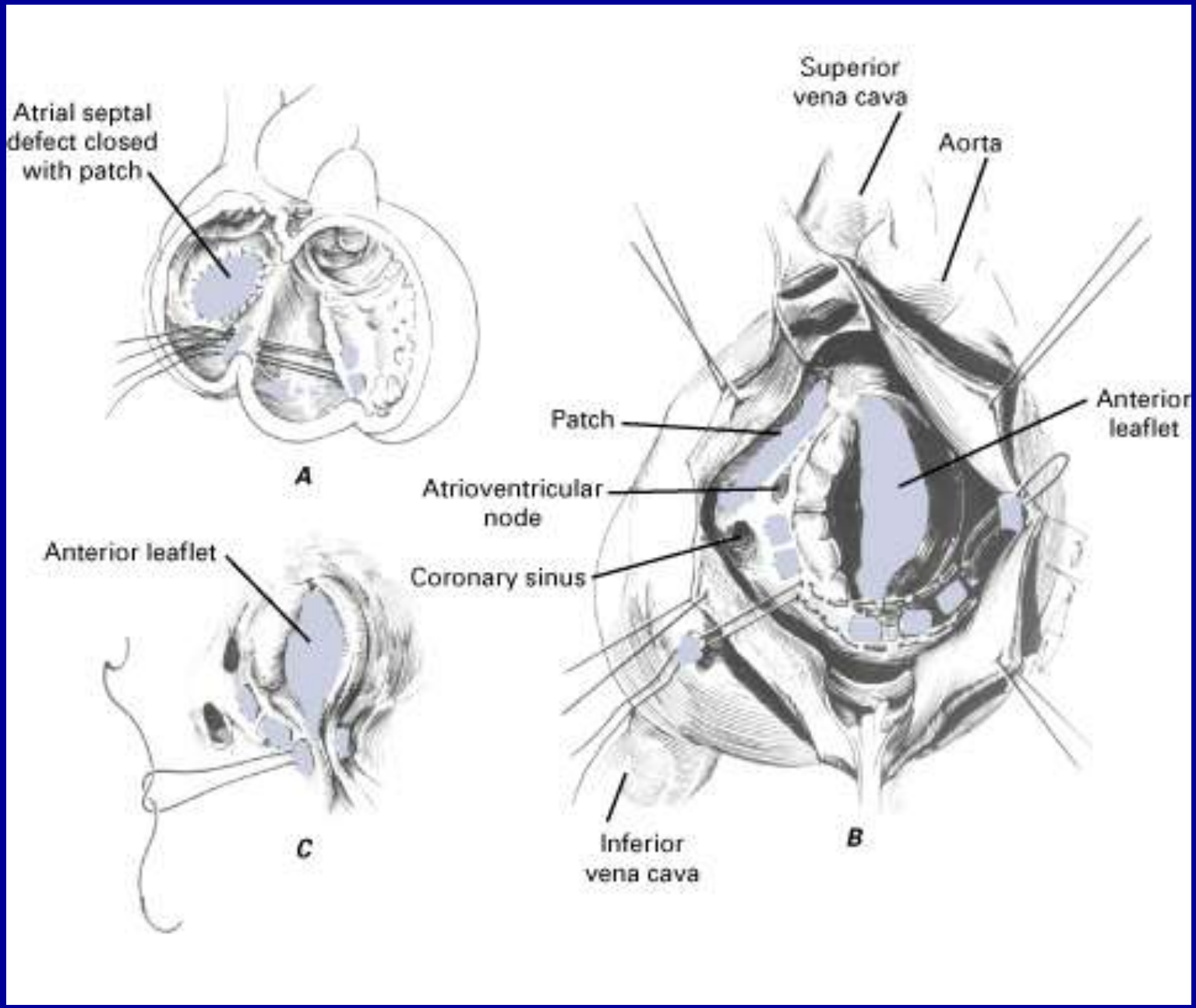


From Hurst, 1999, Ch 70

Ebstein's Anomaly: Arrowheads indicate attachments of TV to IV septum and RV apex



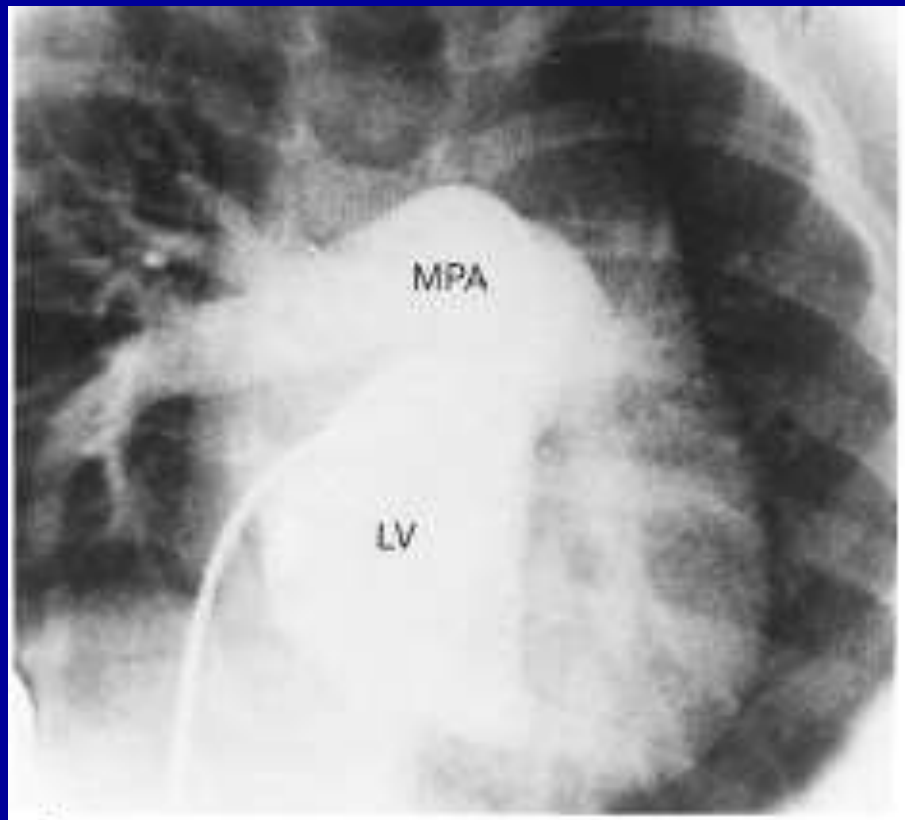
From Hurst, 1999, Ch 70

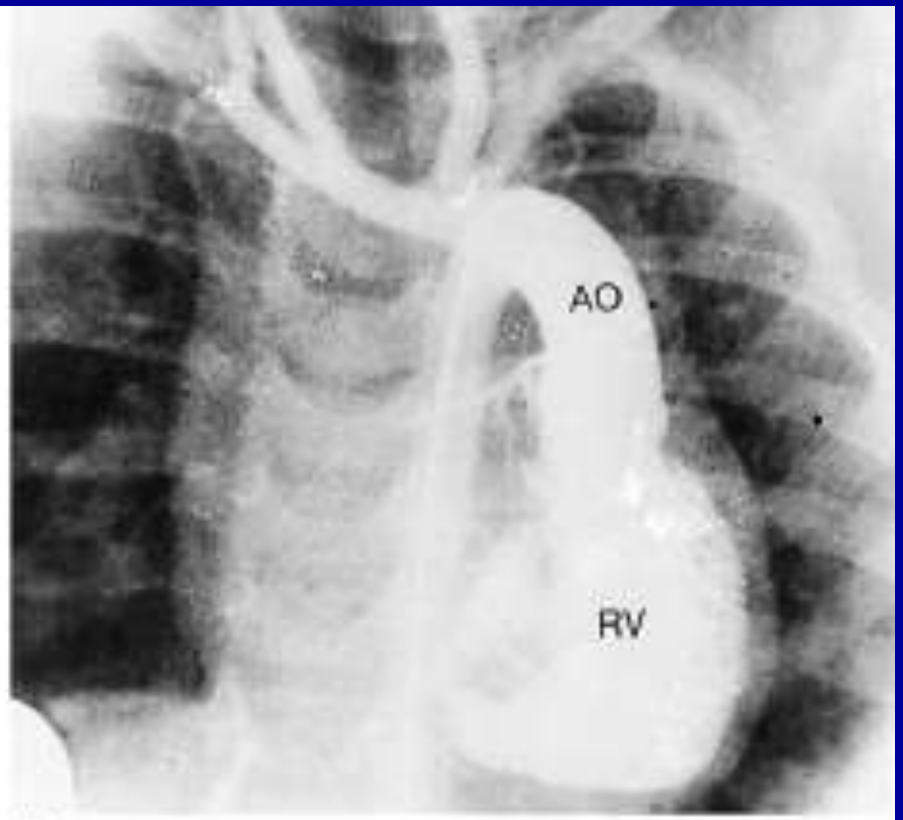


From Hurst, 1999, Ch 70

Repair of Ebstein's Anomaly

Congenitally Corrected Transposition of the Great Arteries

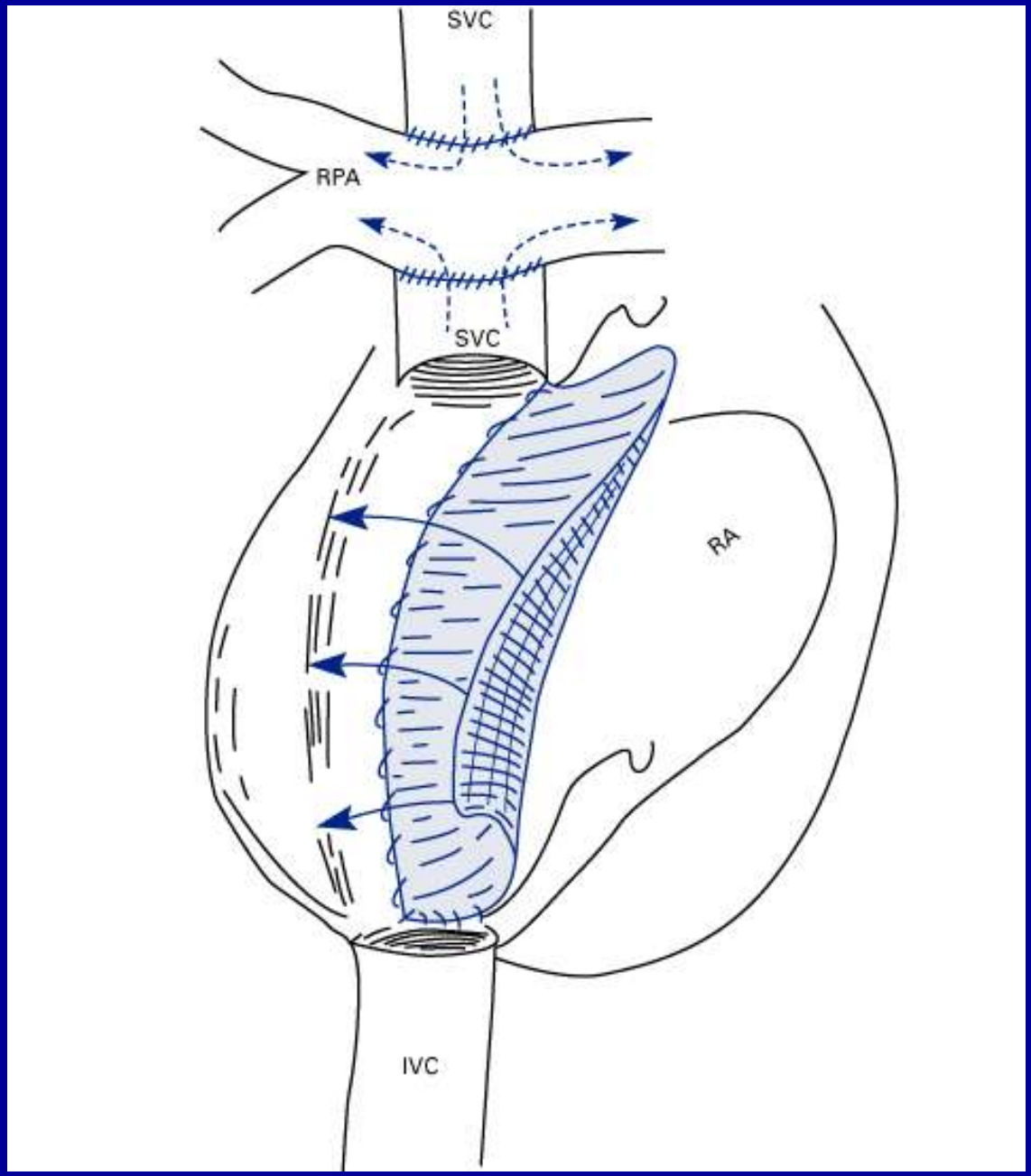




From Hurst, 1999, Ch 70

Modified Fontan Operation

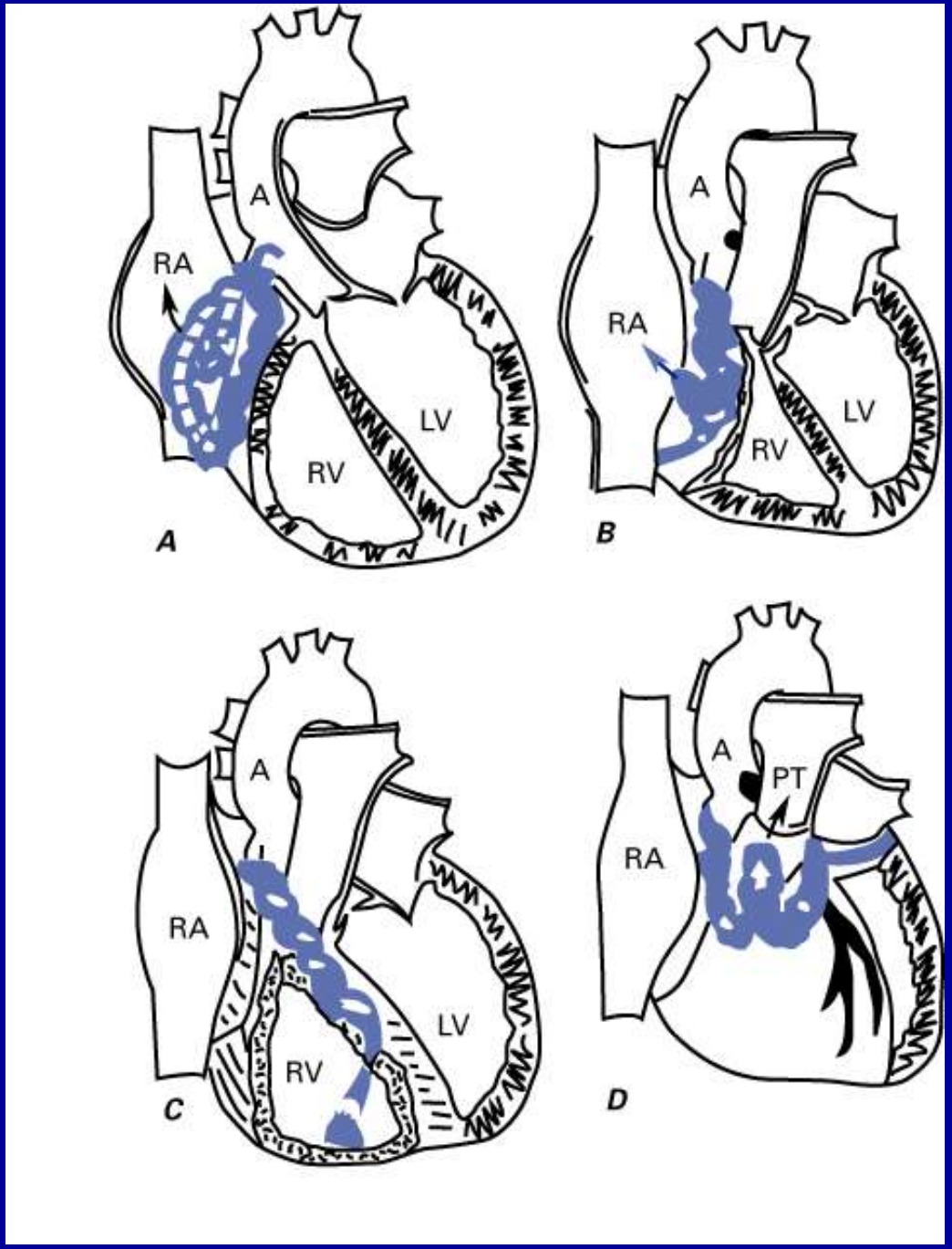
upper SVC to RPA
baffle IVC to SVC
lower SVC to RPA



From Hurst, 1999, Ch 70

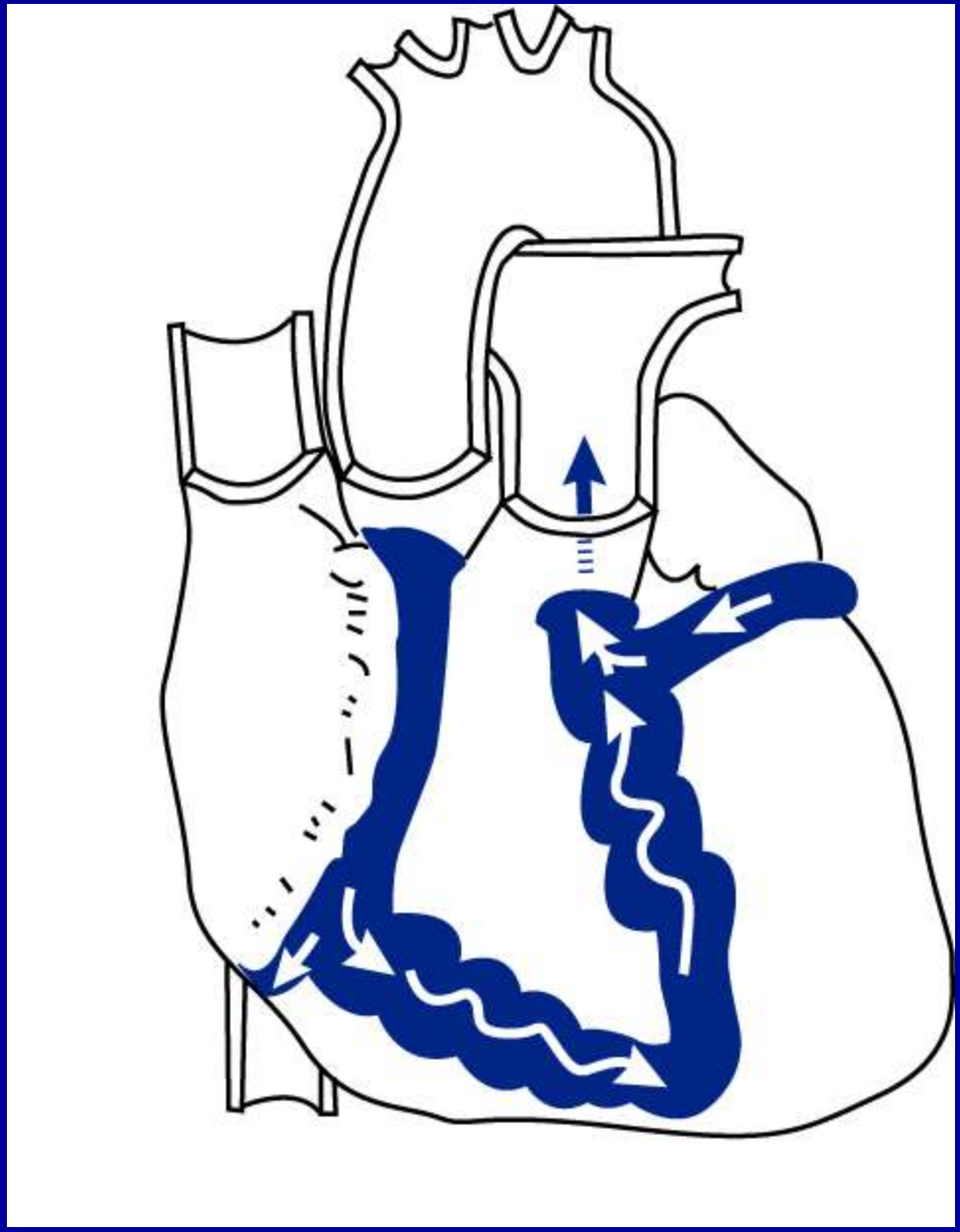
Anomalous Coronary Artery Communications

A: RCA to CS
B: RCA to RA
C: RCA to RV
D: 2 coronary arteries communicate with accessory vessel from PA



From Hurst, 1999, Ch 70

Anomalous Left main coronary From PA



From Hurst, 1999, Ch 70

Coronary Embryology

AO – aorta

PA – pulmonary artery

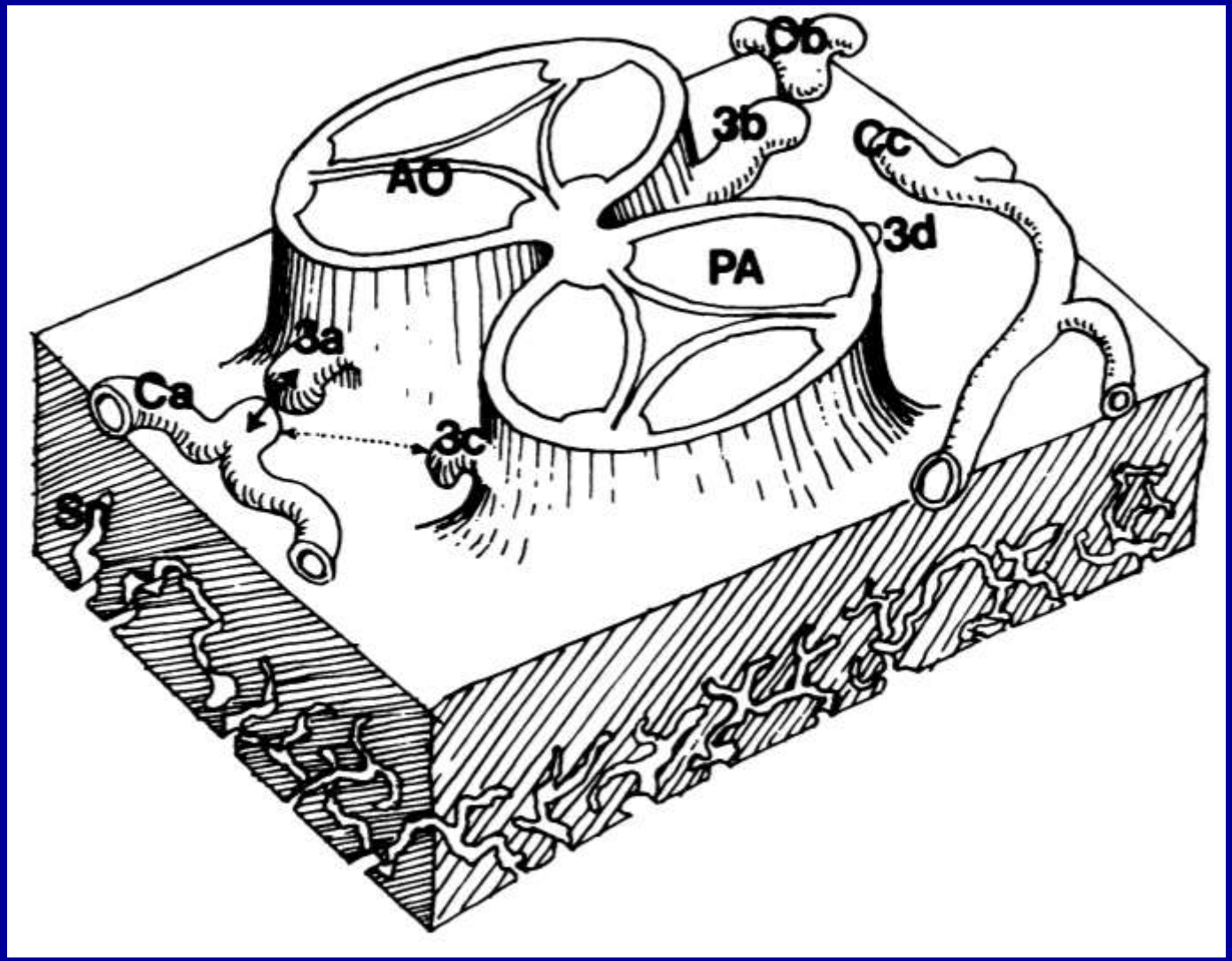
3a-d – coronary buds from semilunar sinuses

Ca – RCA rudiment

Cb – LCX rudiment

Cc – LAD rudiment

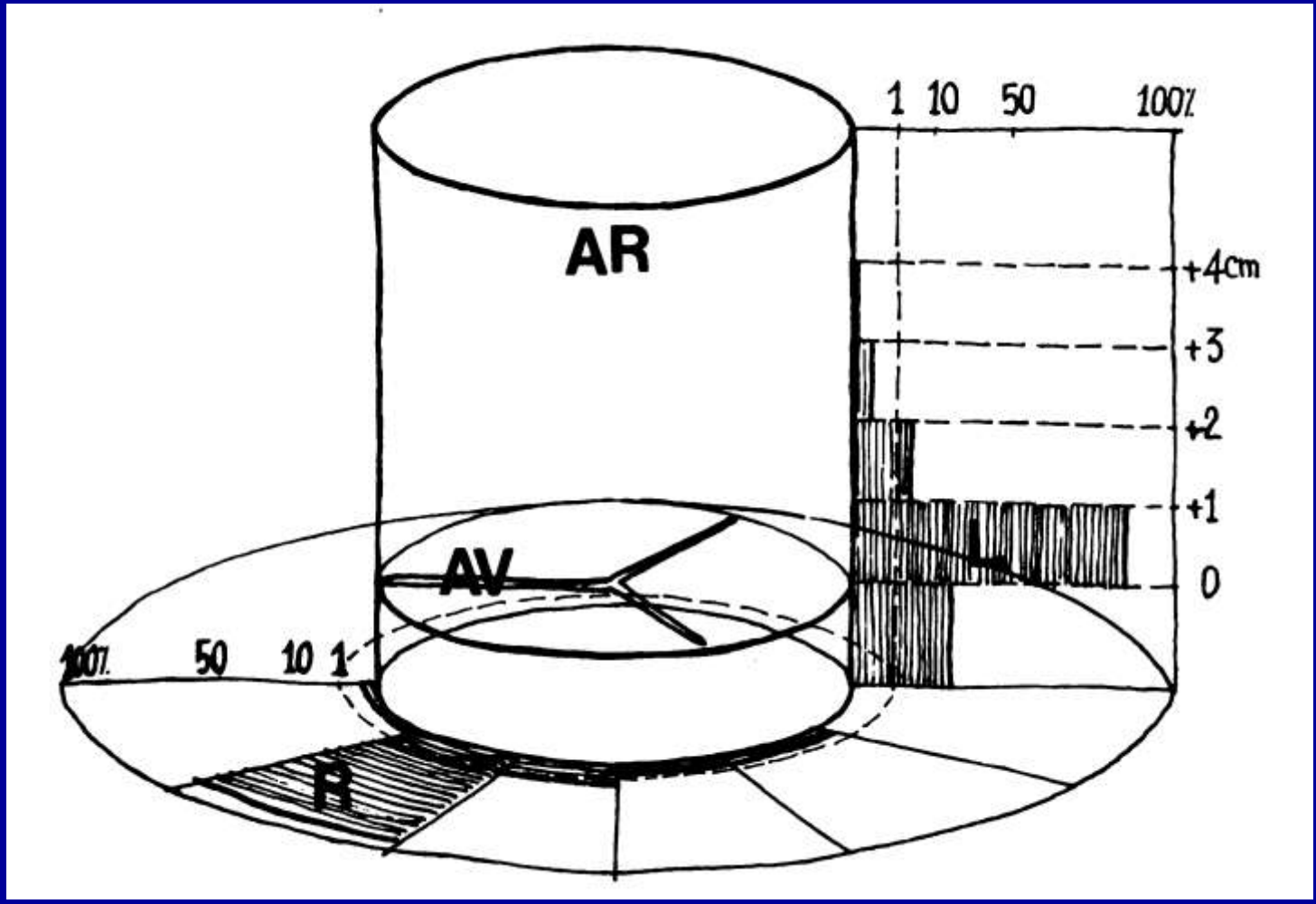
Sn – sinusoids



Angelini P. Am Heart J 1989;117:418

Coronary Ostial Origin Variants

Percentage of variations in coronary ostial sites, with the vertical scale being in cm above (or below) the upper edge of sinus of Valsalva



Angelini P. Am Heart J 1989;117:418

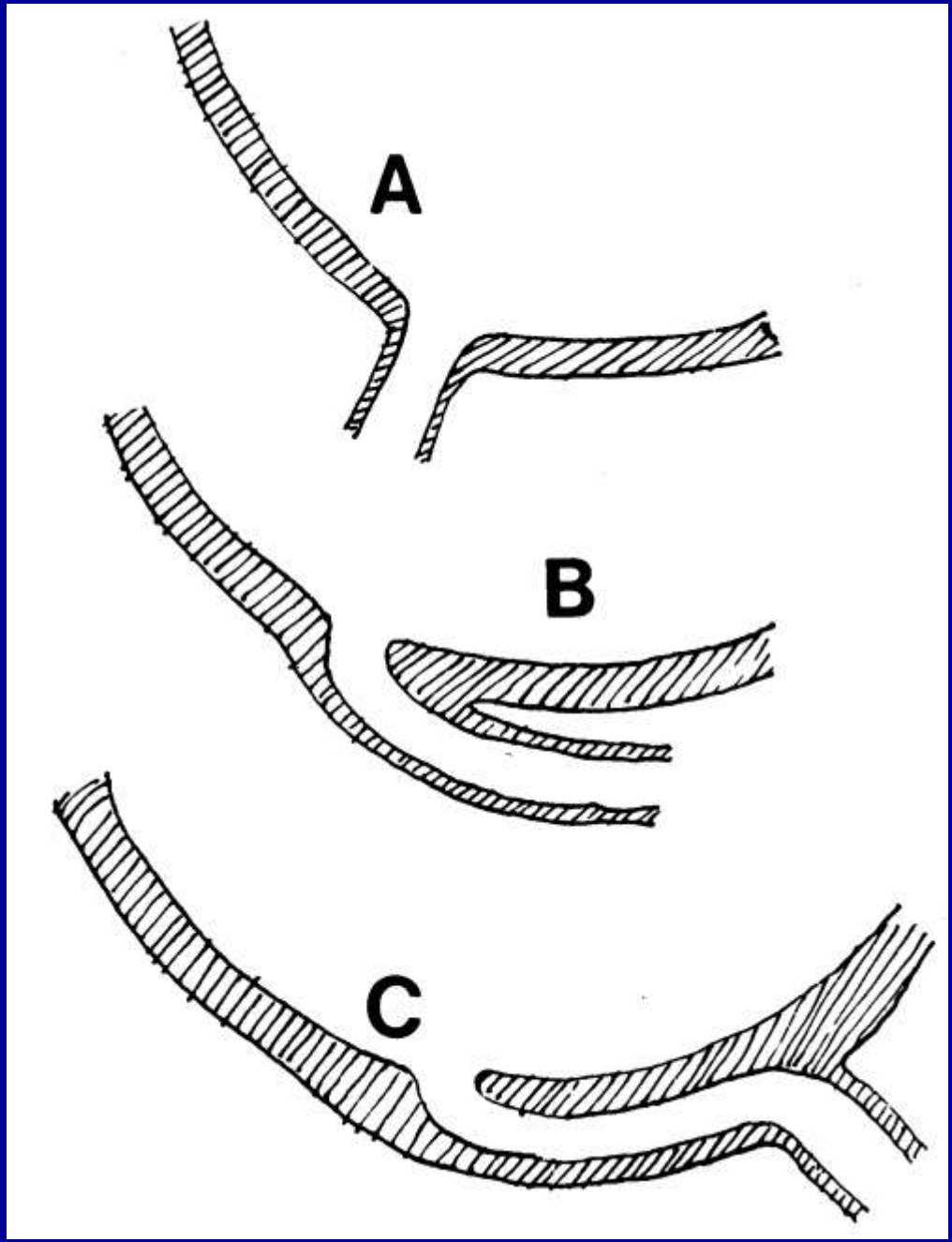
Coronary Ostial Origin Variants

Variations in coronary ostial orientation

A – common, coronary is nearly orthogonal to aortic wall in both vertical and horizontal axes

B – less frequent, nearly tangential

C – unusual, intussusception of coronary artery, proximal segment is embedded in aortic wall



Angelini P. Am Heart J 1989;117:418

Variants in Origin and Course

I – posterior

II – retroaortic

III – intertruncal

IV – intramuscular (within crista supraventricularis and ventricular septum)

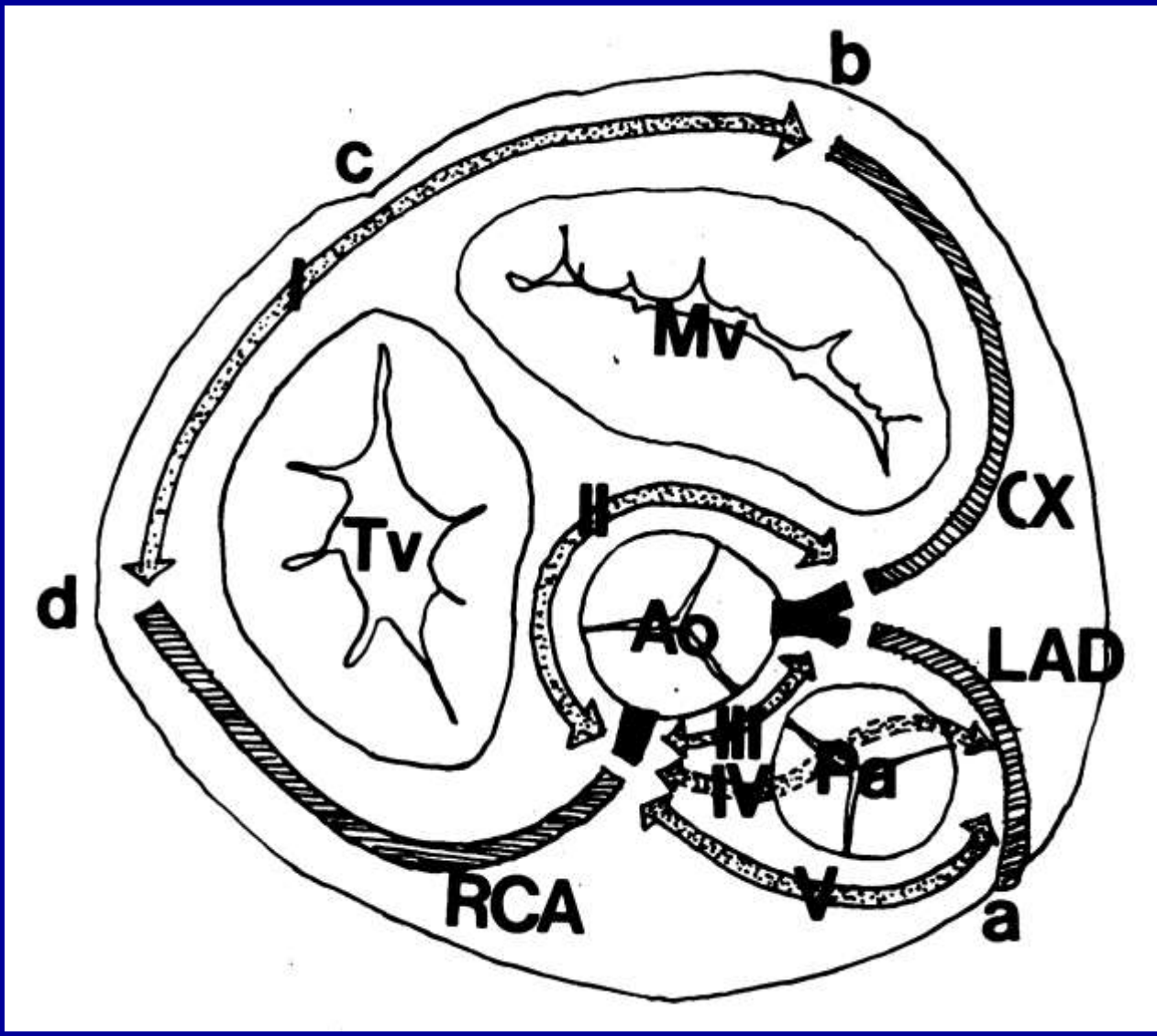
V – anterior (within pulmonary infundibulum)

a: anterior interventricular sulcus

b: obtuse cardiac margin

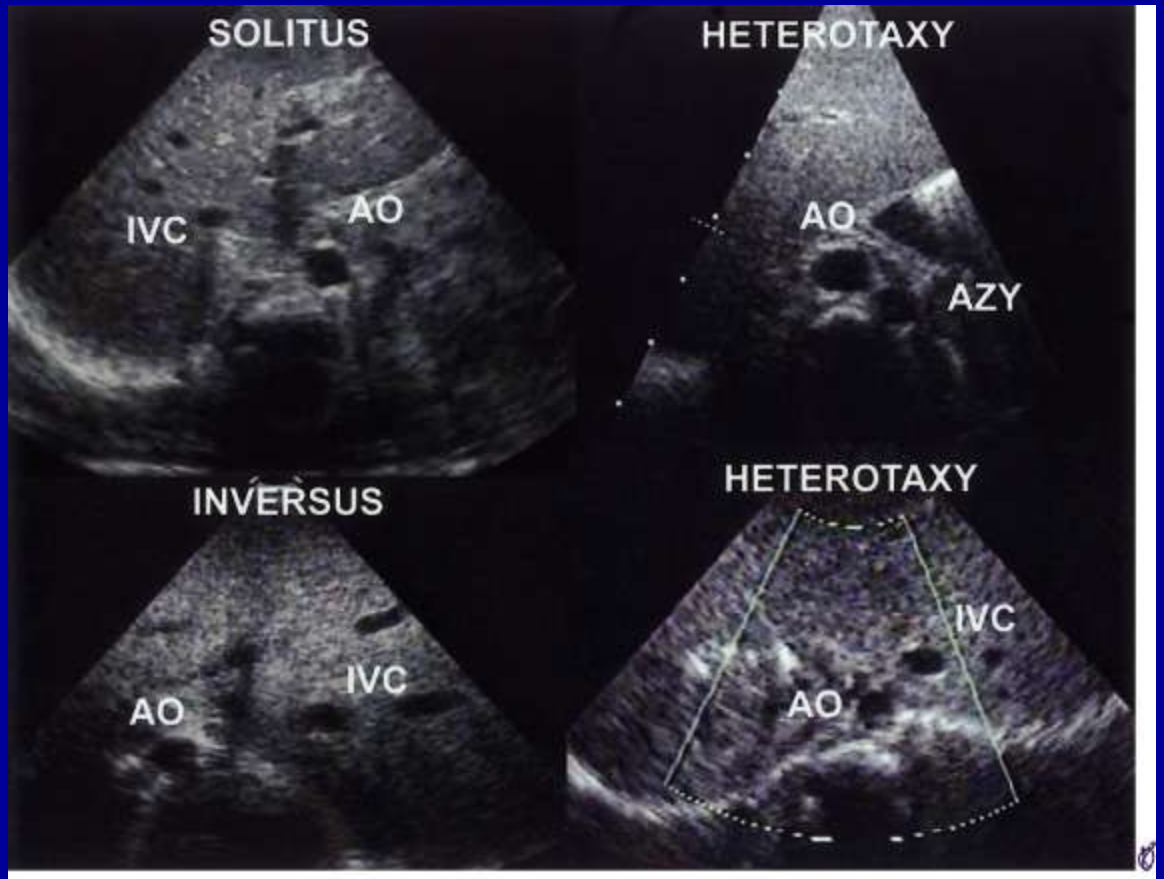
c: acute cardiac margin

d: posterior interventricular sulcus

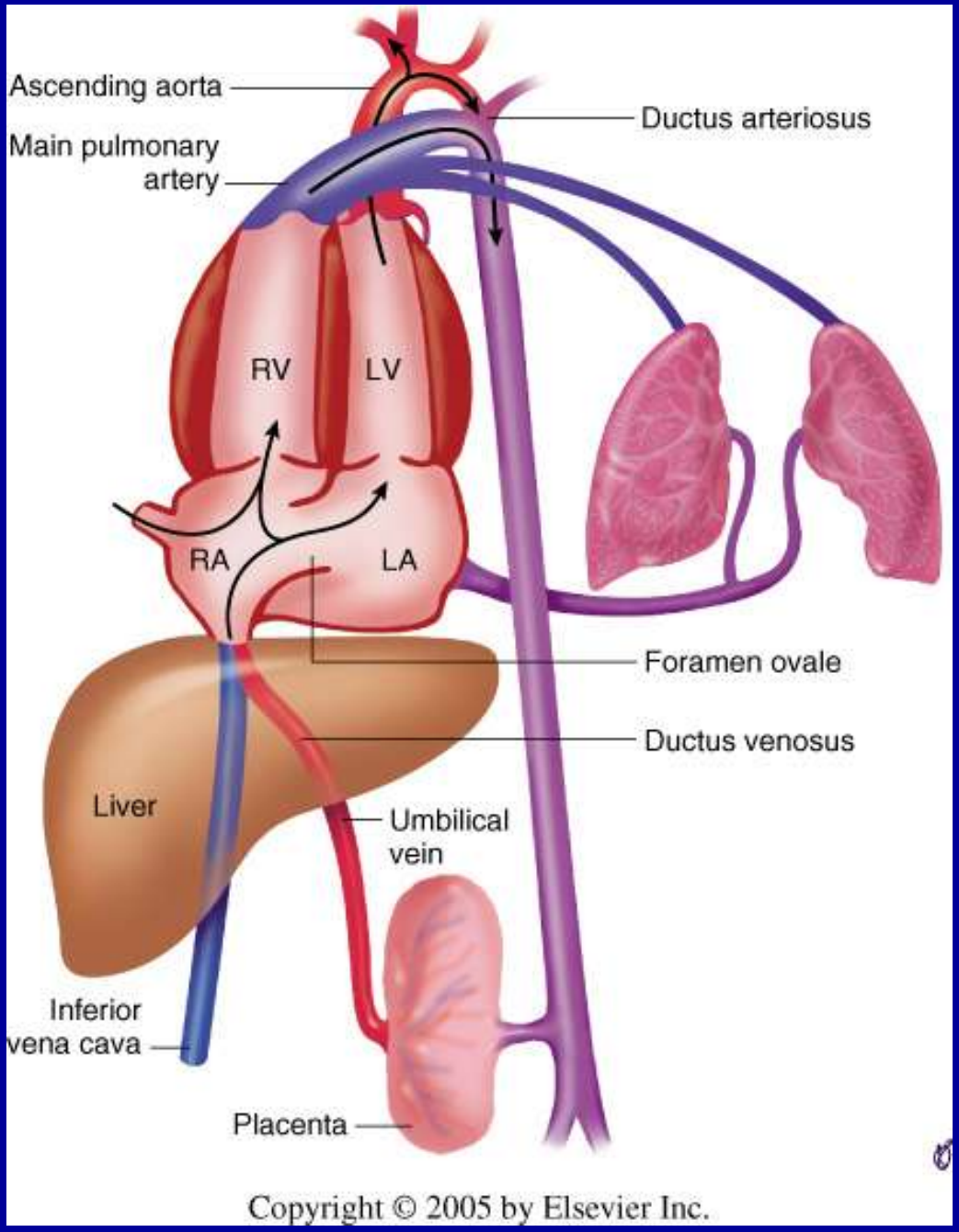


Angelini P. Am Heart J 1989;117:418

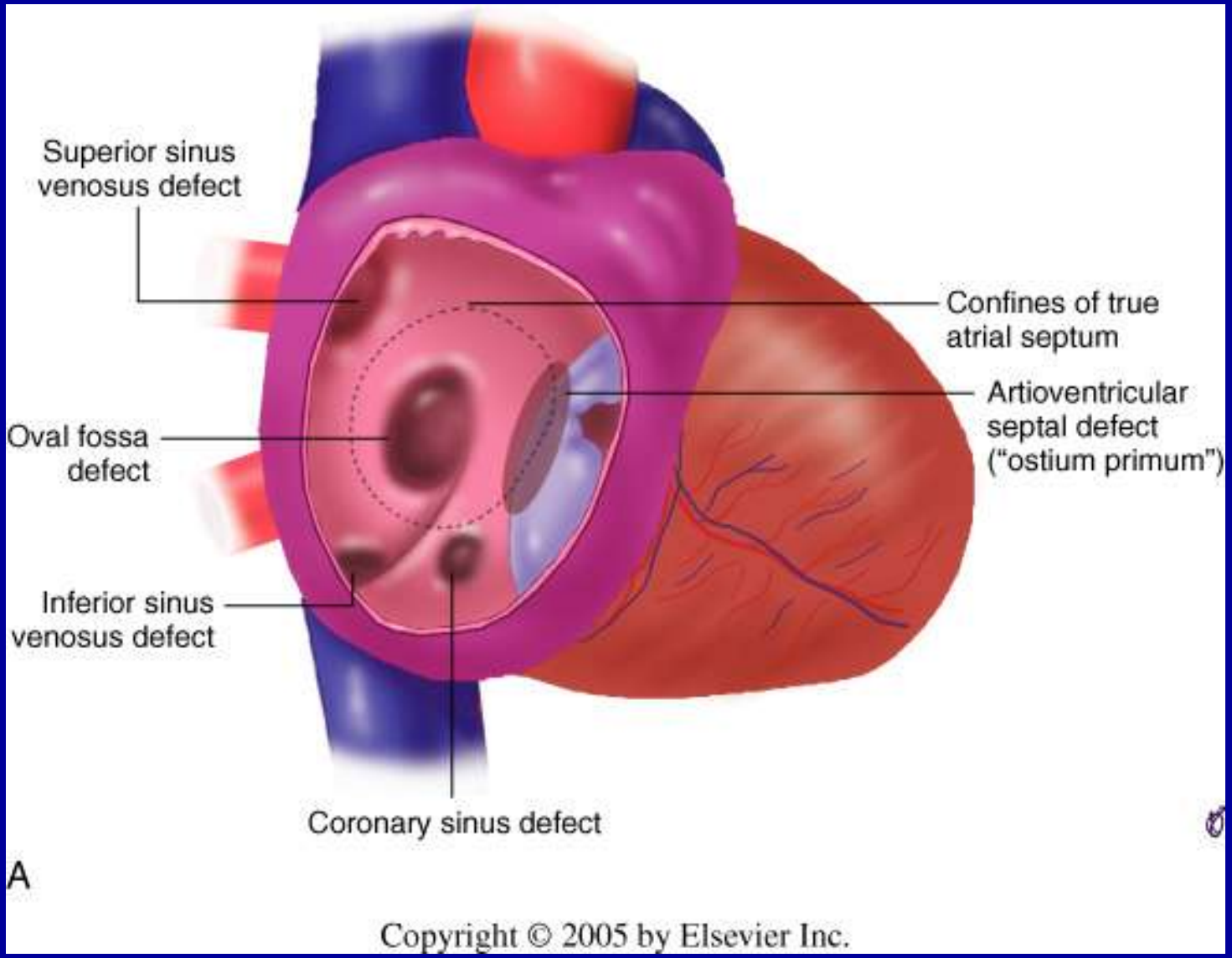
Situs Types by Subcostal Echo



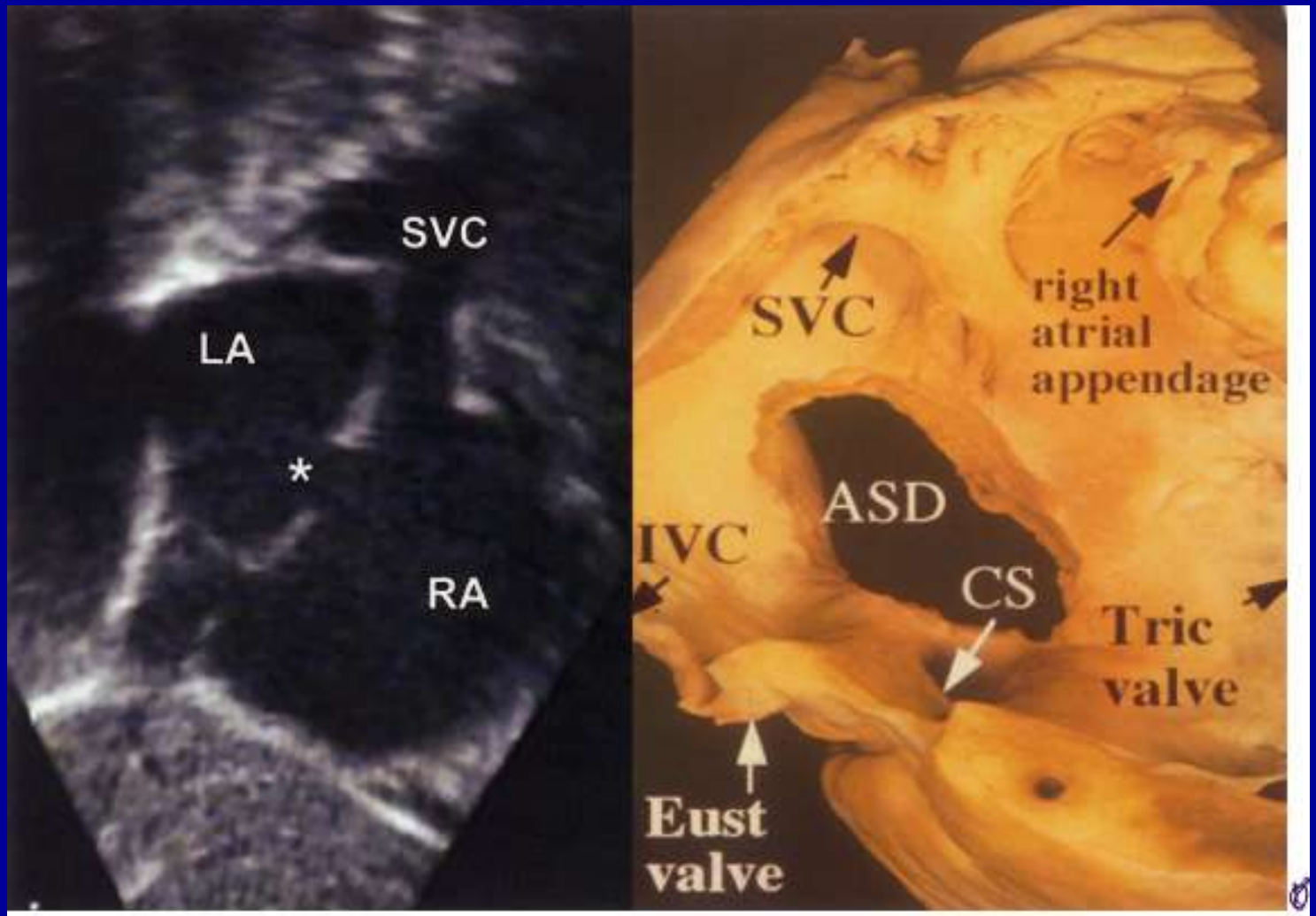
Fetal Circulation



Types of ASD

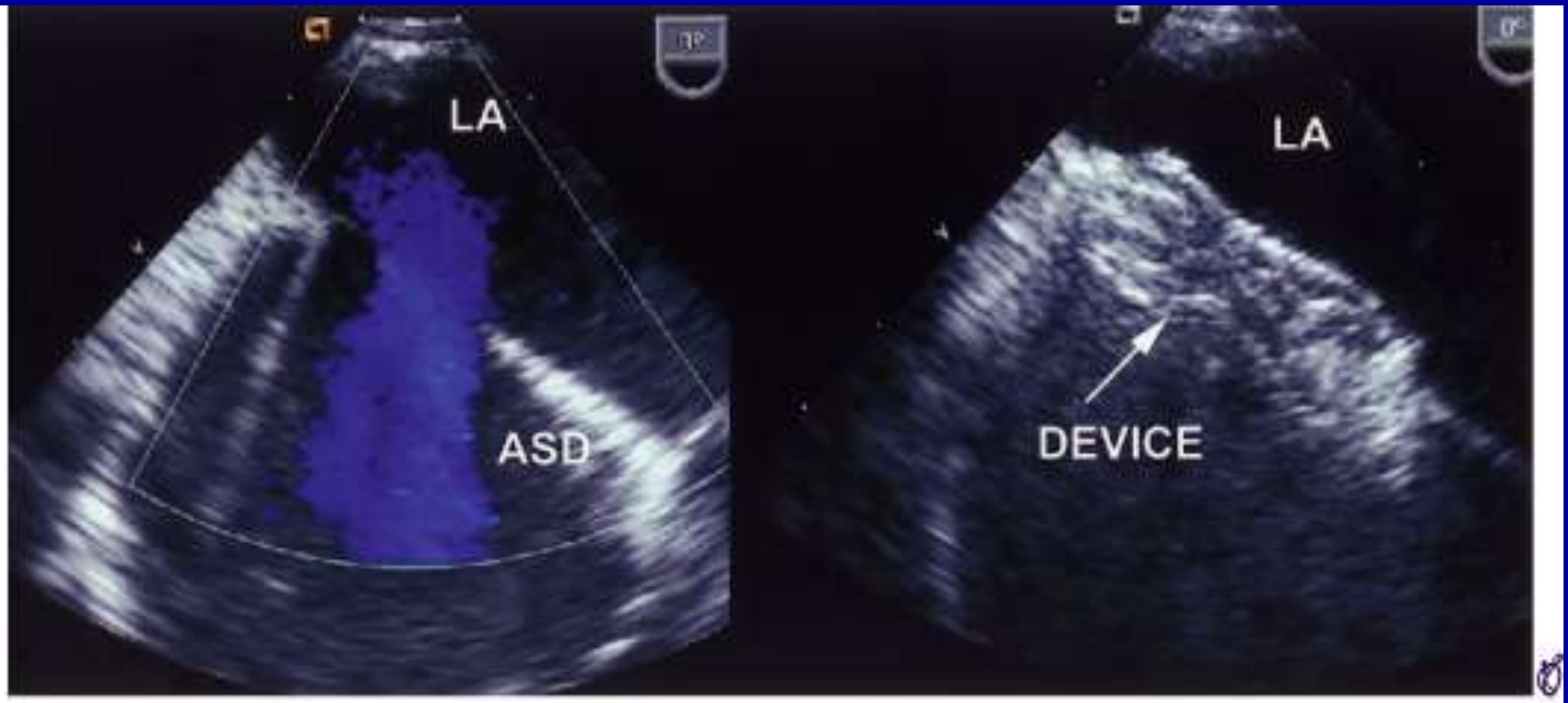


Secundum ASD (subcostal RAO view)



B

Secundum ASD and Amplatzer Closure



C

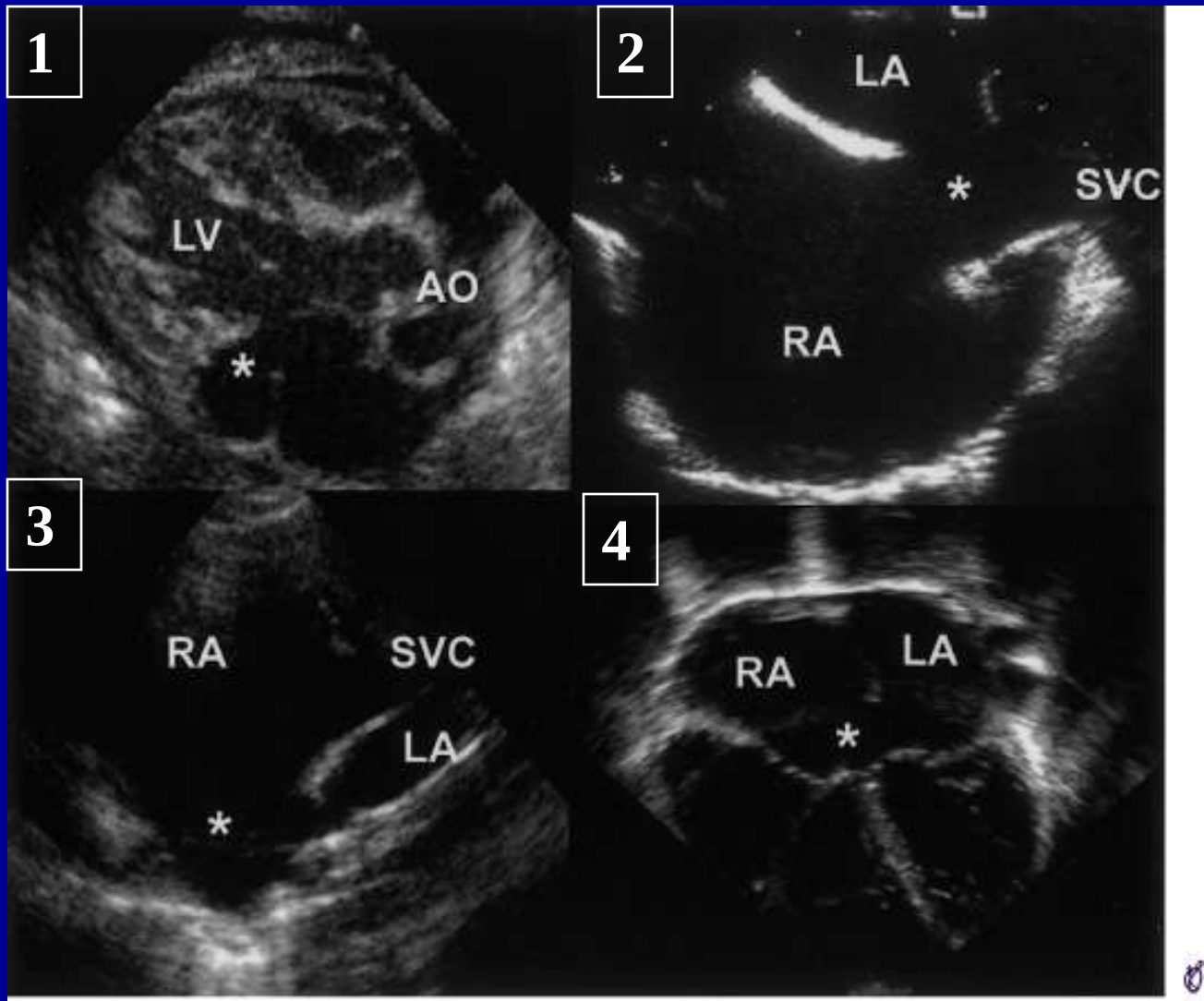
Other Interatrial Communications

1. Coronary sinus defect due to unroofing

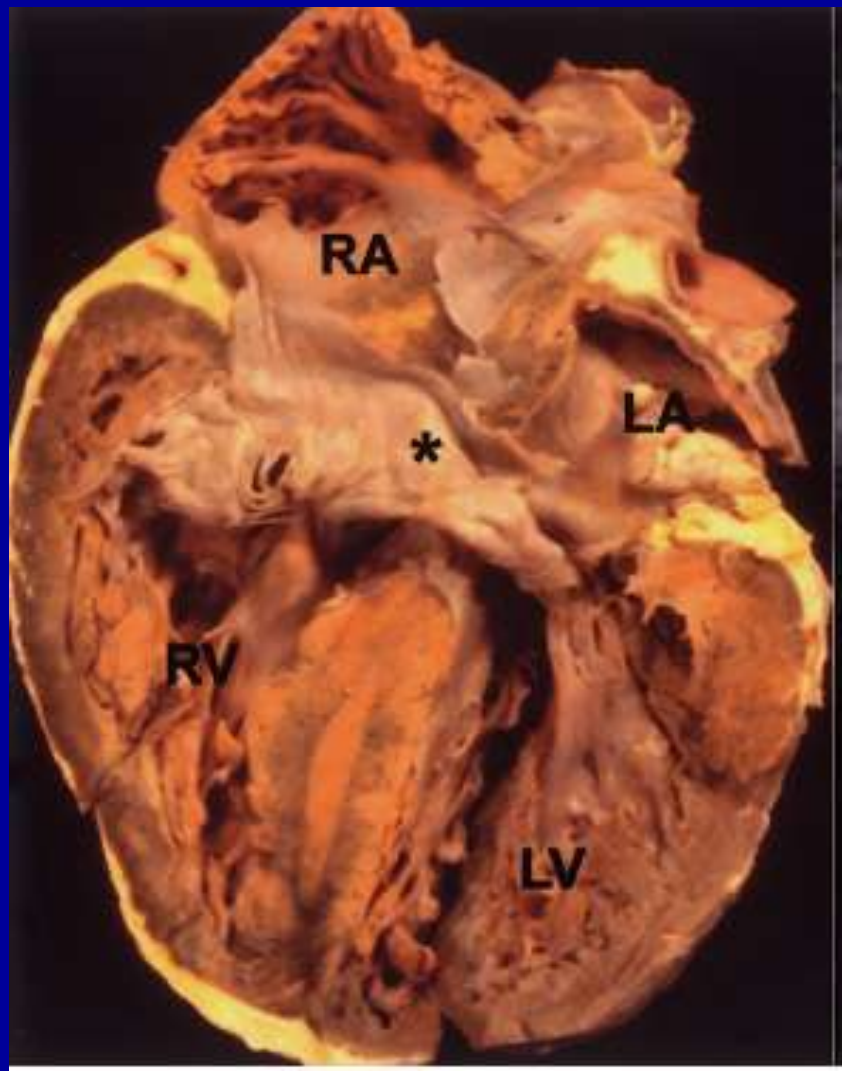
2. Superior sinus venosus defect

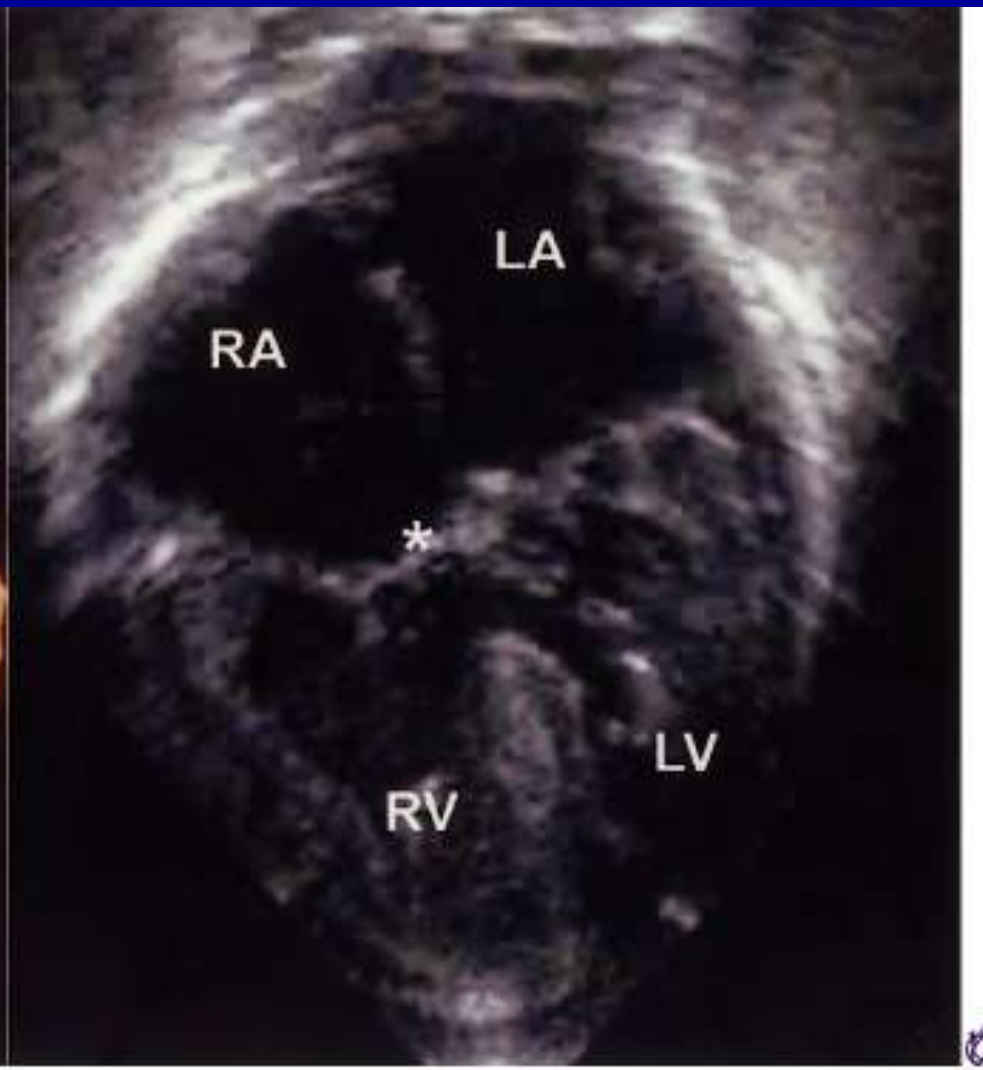
3. Inferior sinus venosus defect

4. Atrioventricular septal defect

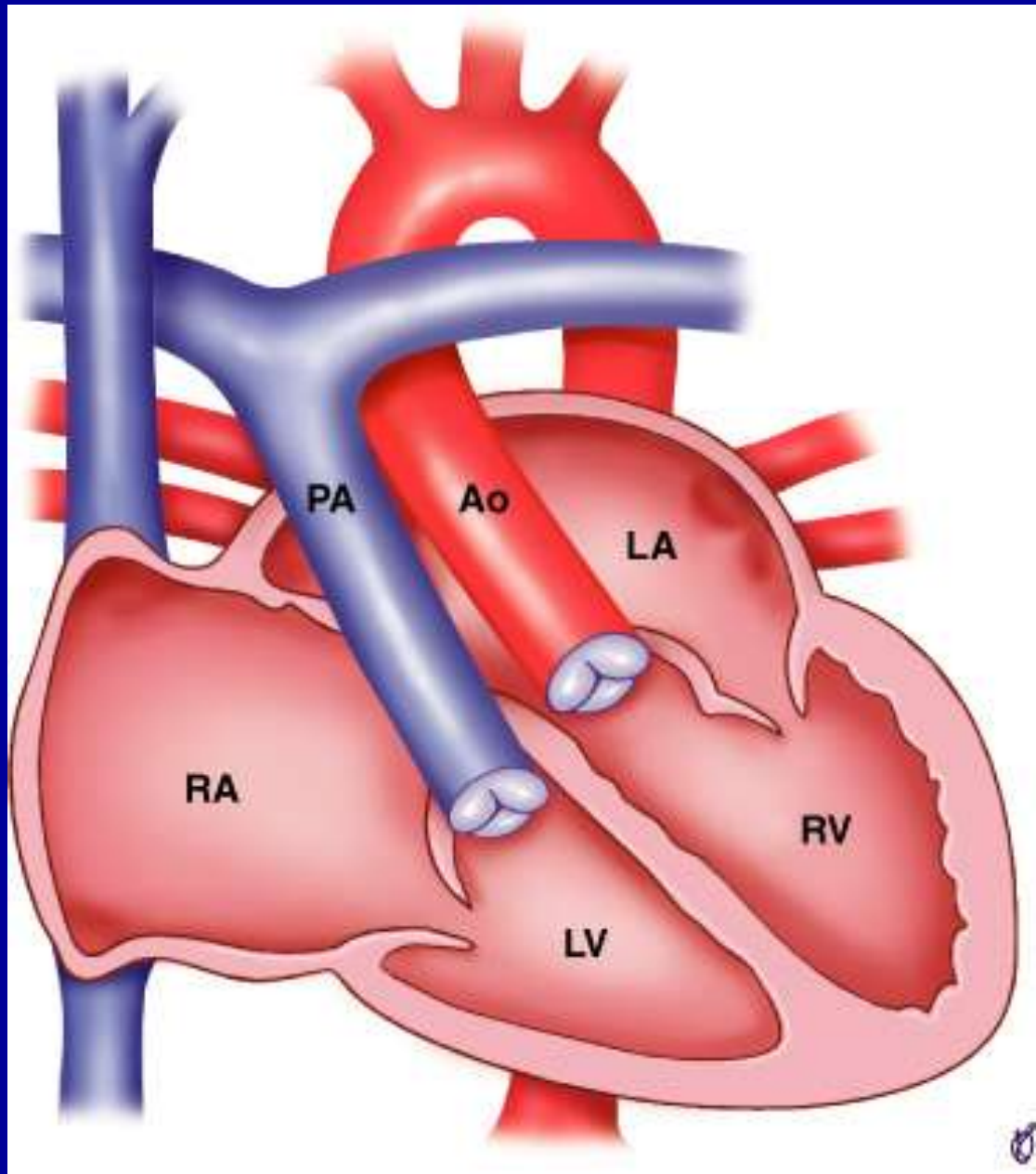


Complete Atrioventricular Septal Defect

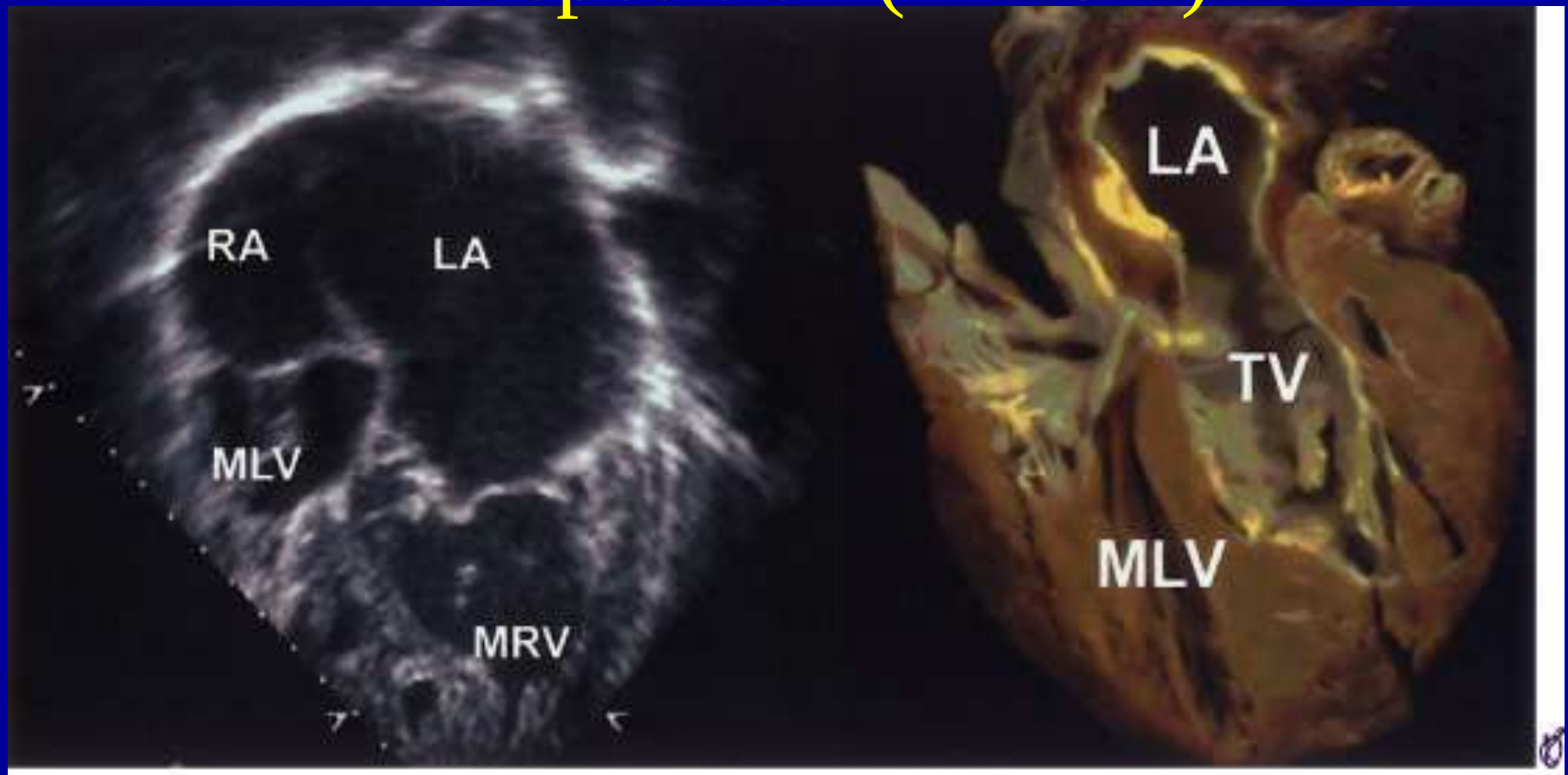




Congenitally Corrected Transposition (L-TGA)

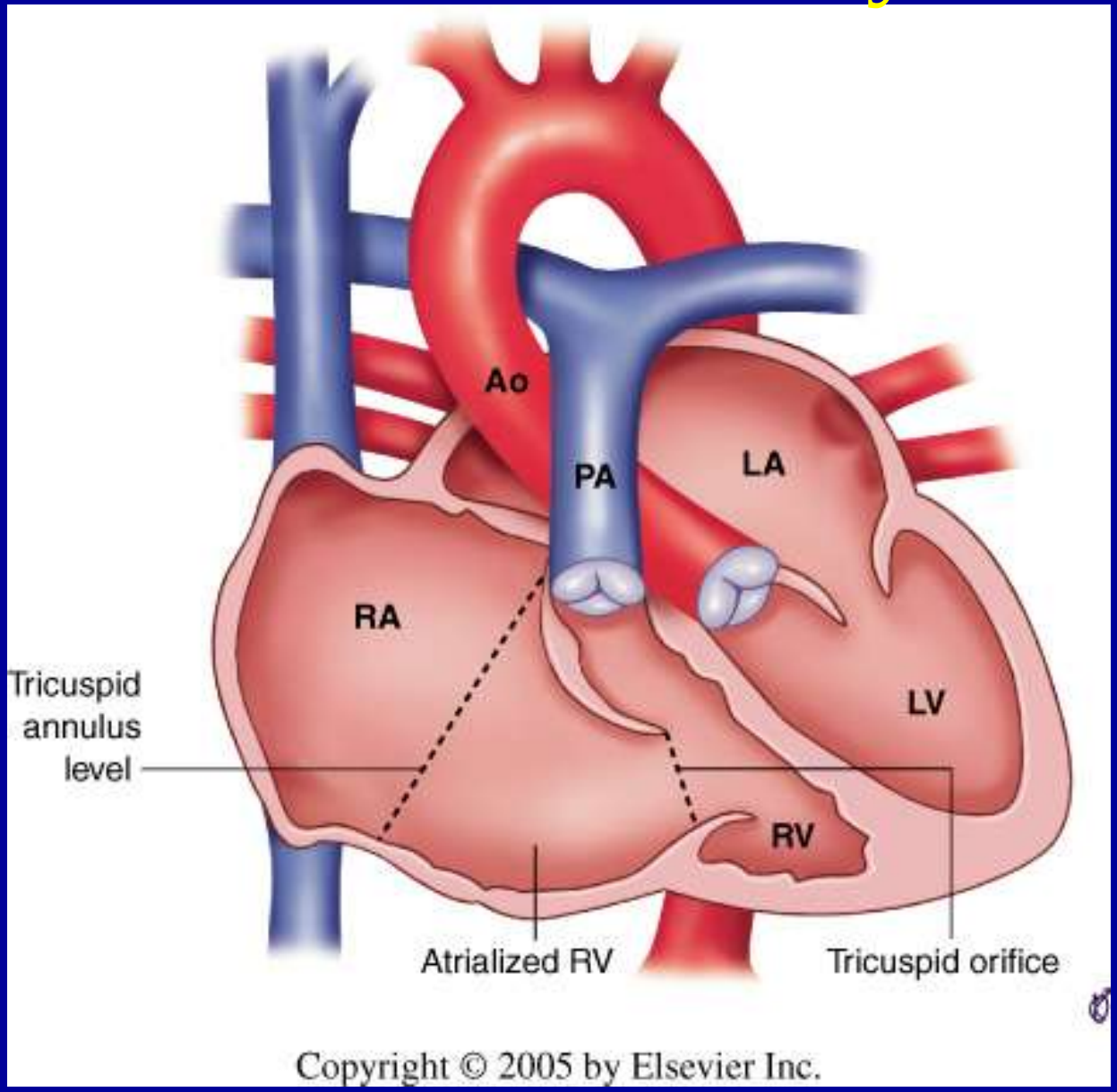


Congenitally Corrected Transposition (L-TGA)

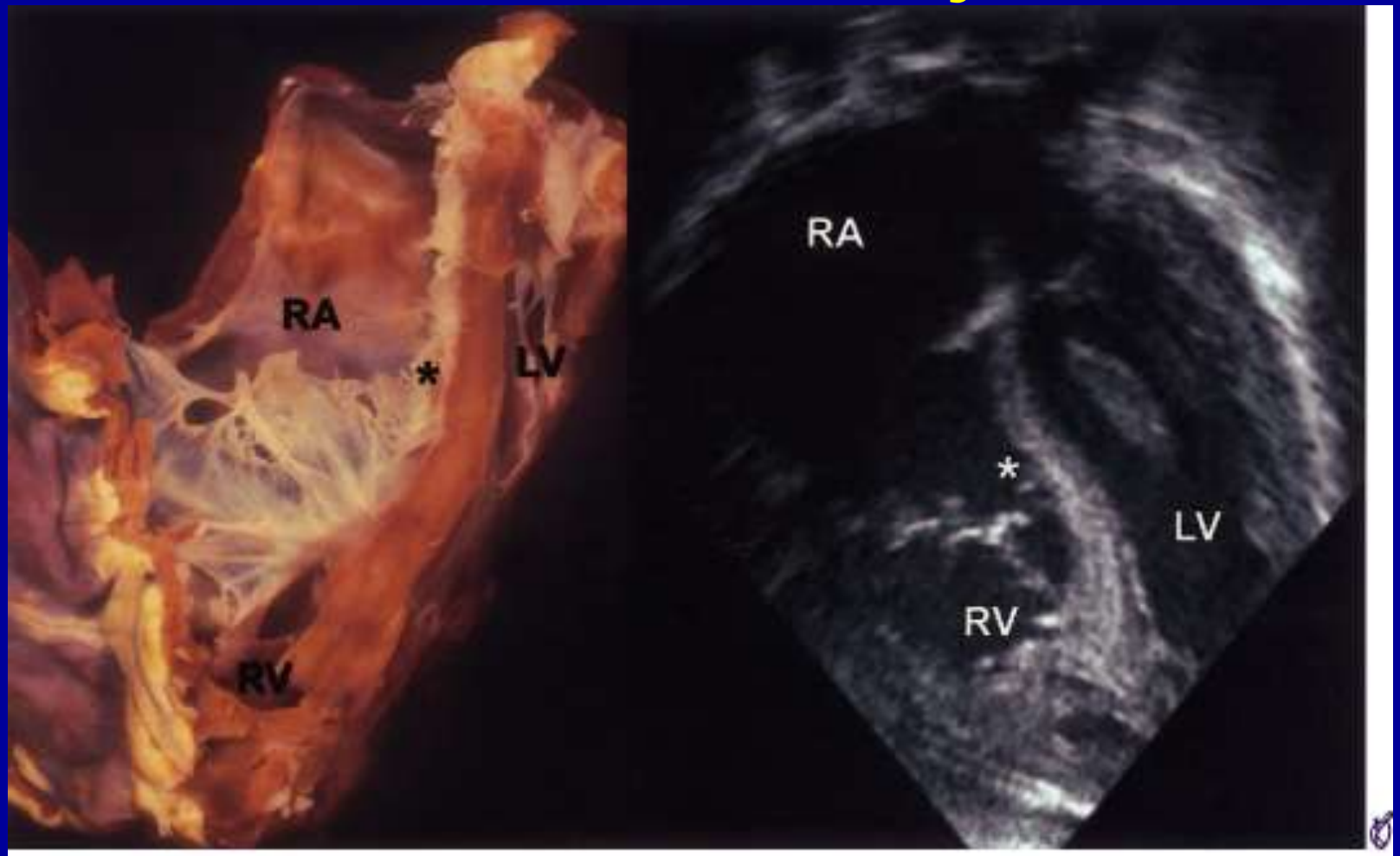


dysplasia and displacement of the morphological left-sided tricuspid valve

Ebstein anomaly

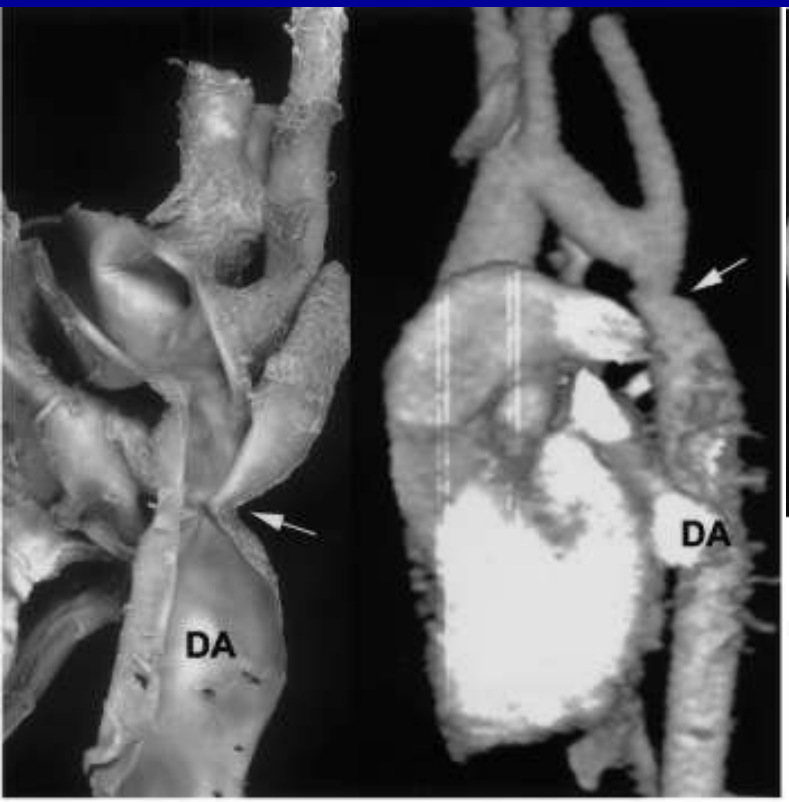


Ebstein anomaly



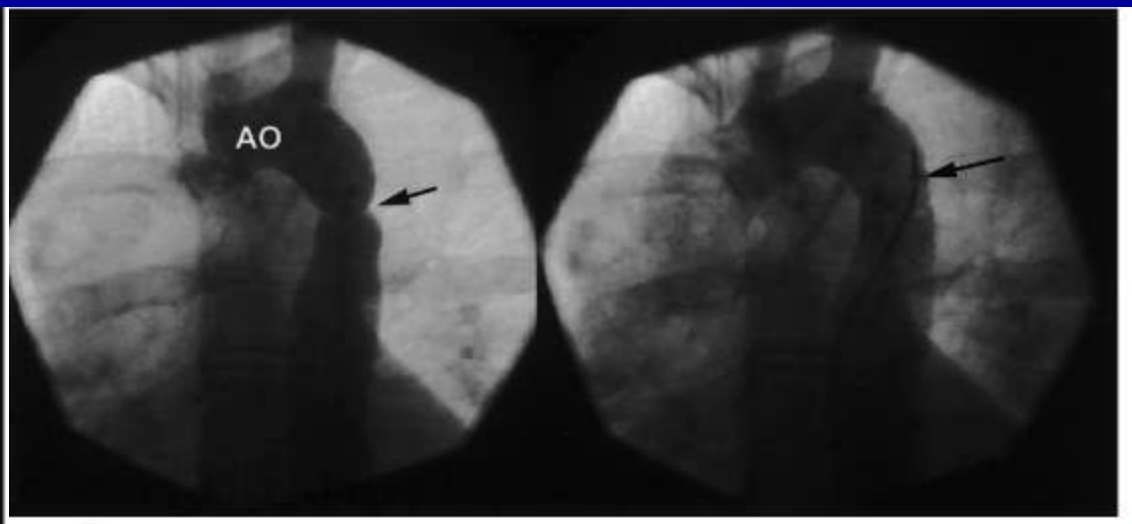
significant displacement of the septal tricuspid valve leaflet (asterisk), with associated valve dysplasia

Coarctation of the Aorta



A

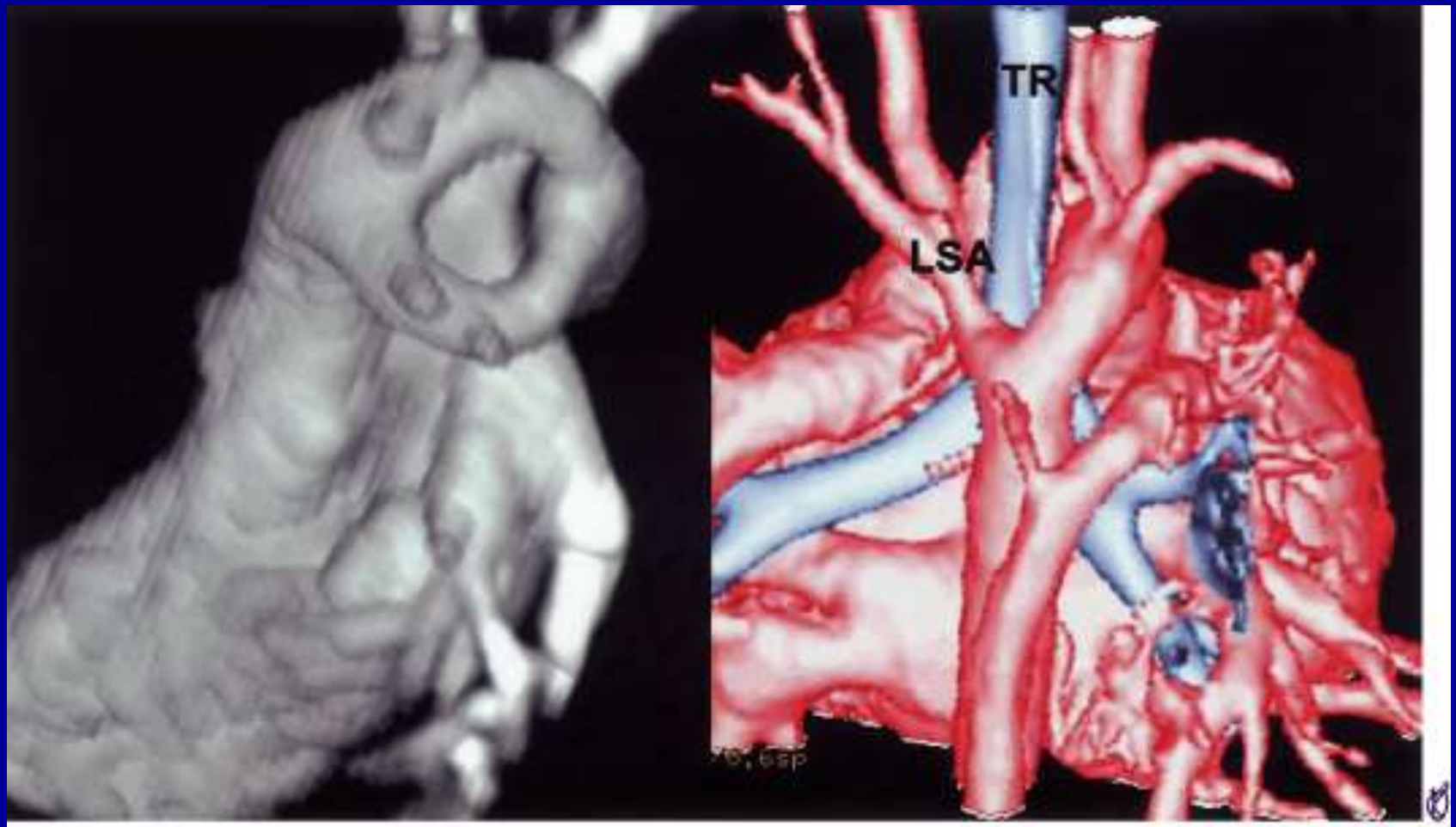
Site of the posterior shelf, as outlined by the arrow



B

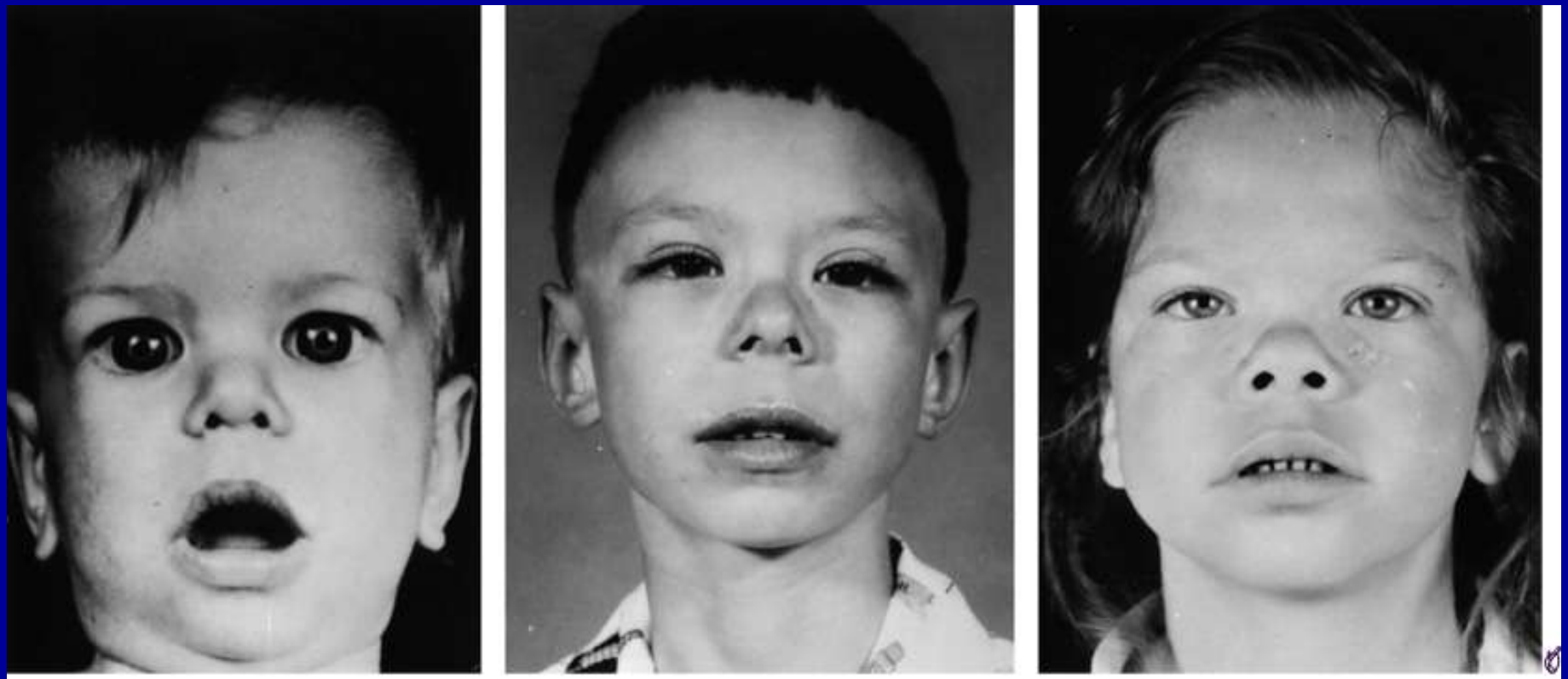
Coarctation of the aorta, before and after stenting

Double Aortic Arch



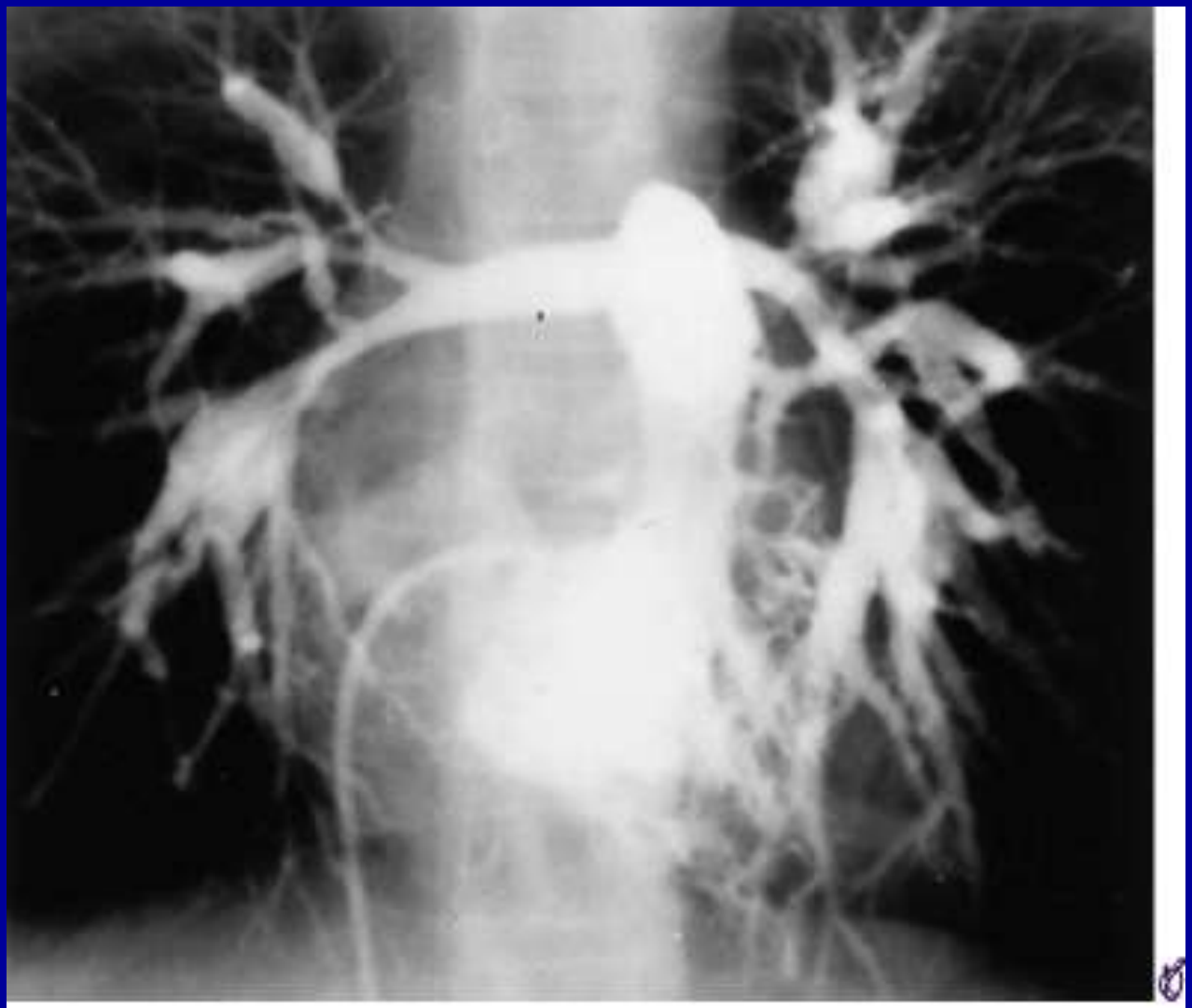
The right image is from an aberrant left subclavian artery as seen by spiral CT

Supravalvular Aortic Stenosis



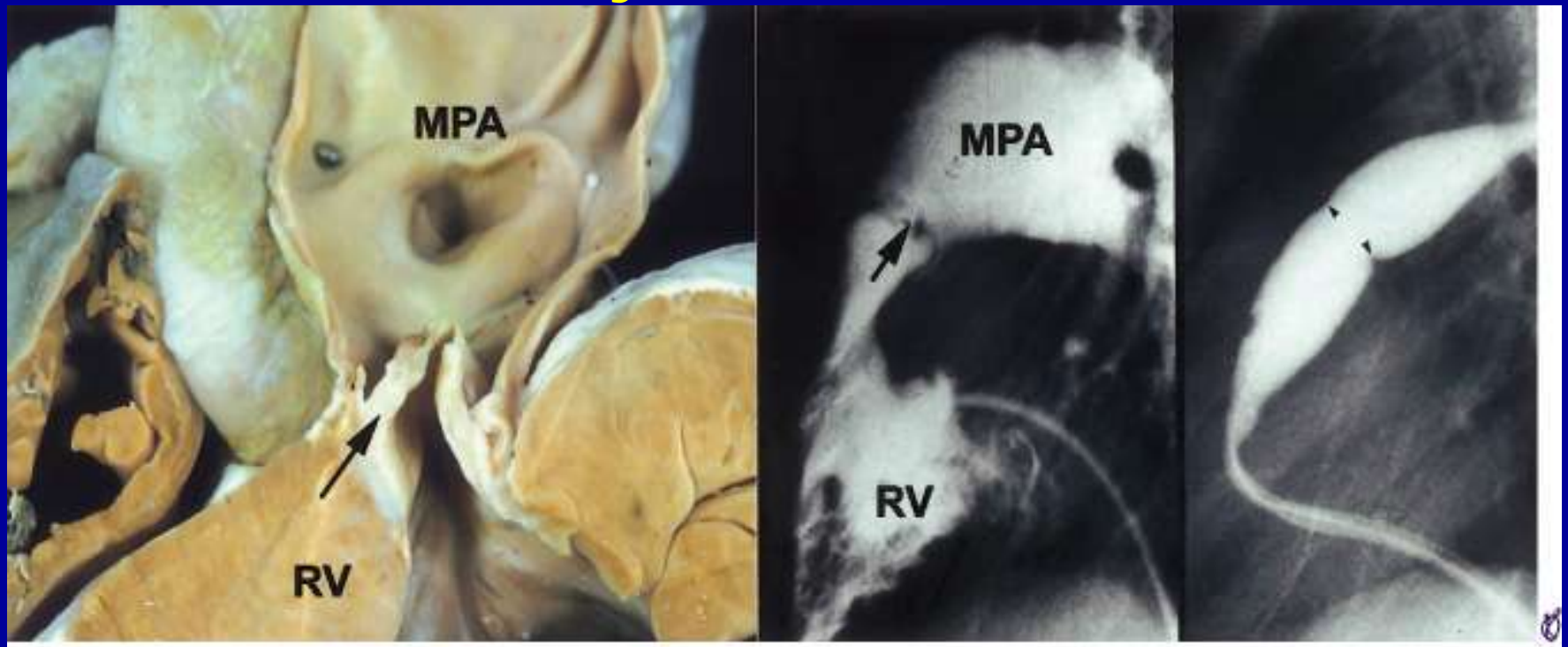
“Elfin facies”

Peripheral Pulmonic Stenosis



poststenotic dilation of the peripheral pulmonic arteries

Pulmonary Valve Stenosis

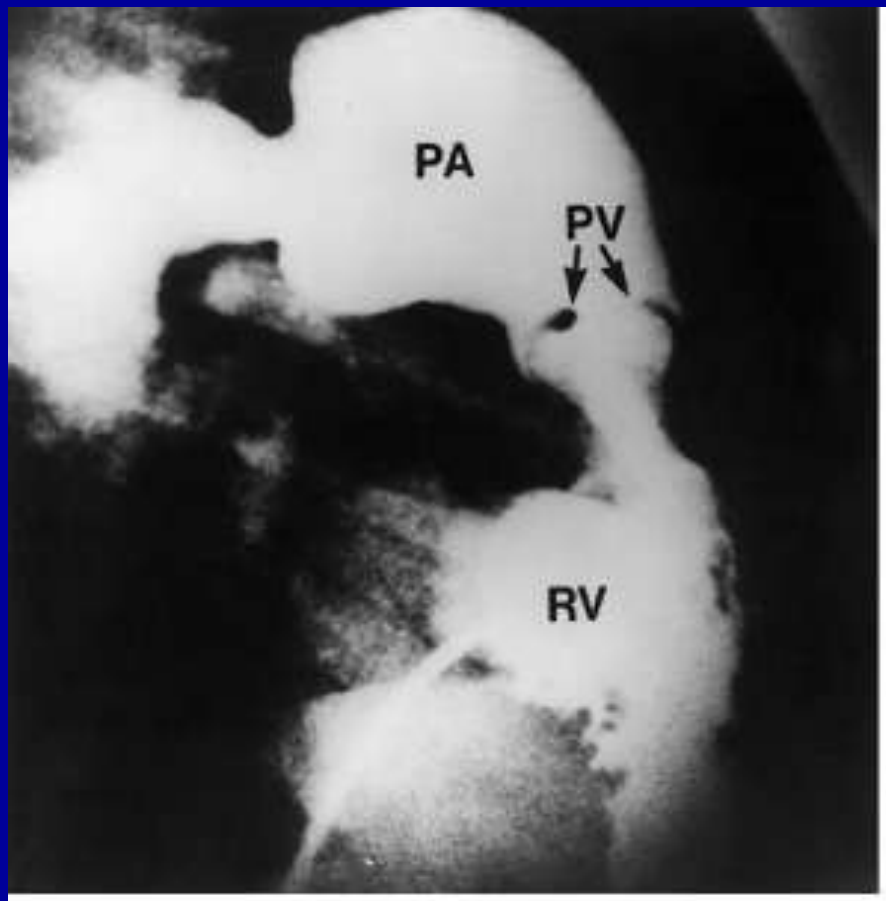


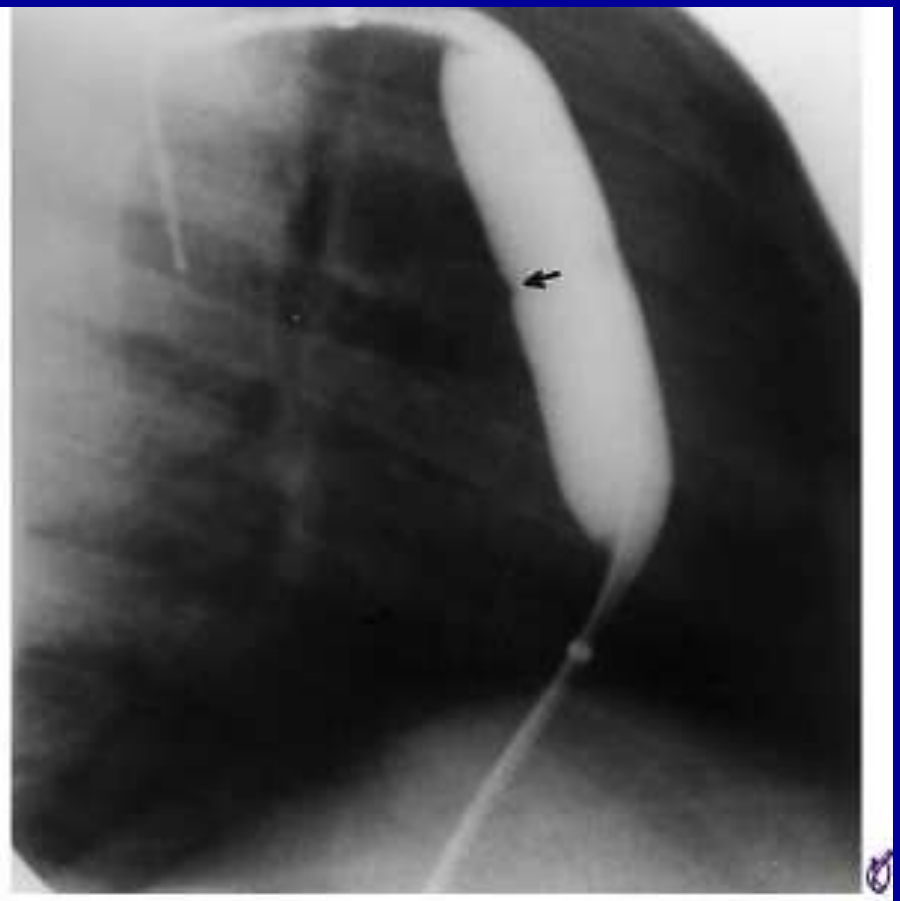
a. Thickened pulmonary valve and obstruction due to commissural fusion

b. Post-stenotic dilation

c. Balloon dilation

Pulmonary Valve Stenosis





Pulmonary Vein Stenosis

Three-dimensional MRI demonstrating stenosis of the left lower lobe pulmonary vein