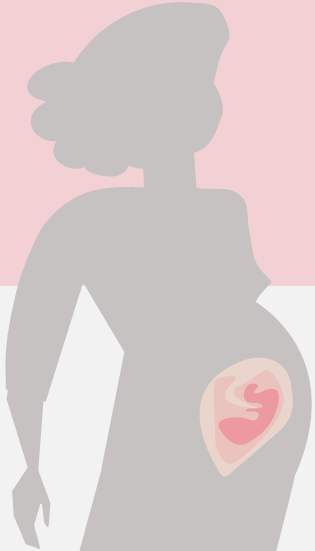
Prenatal ultrasound - ASD, VSD



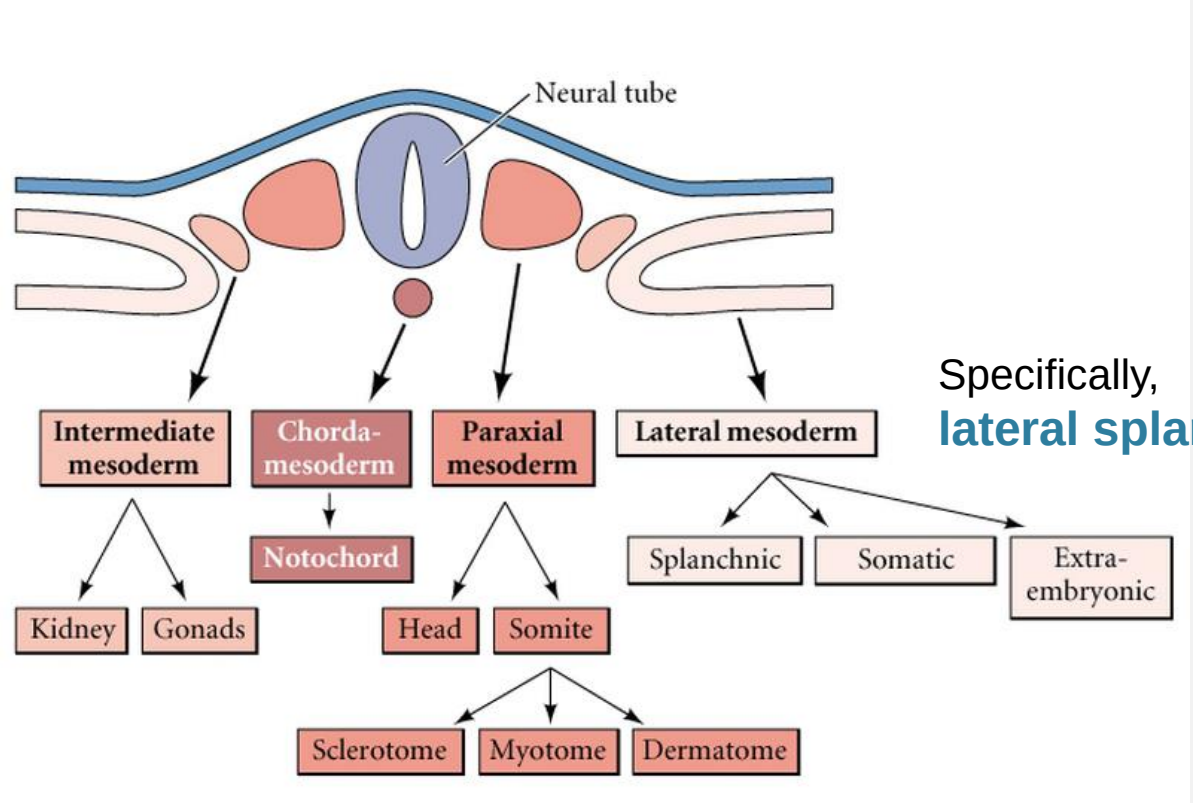
Presenter: PGY2 張家甄
Supervisor: VS 林宜珈醫師

2022/05/06

Outline

Anatomy, Embryology

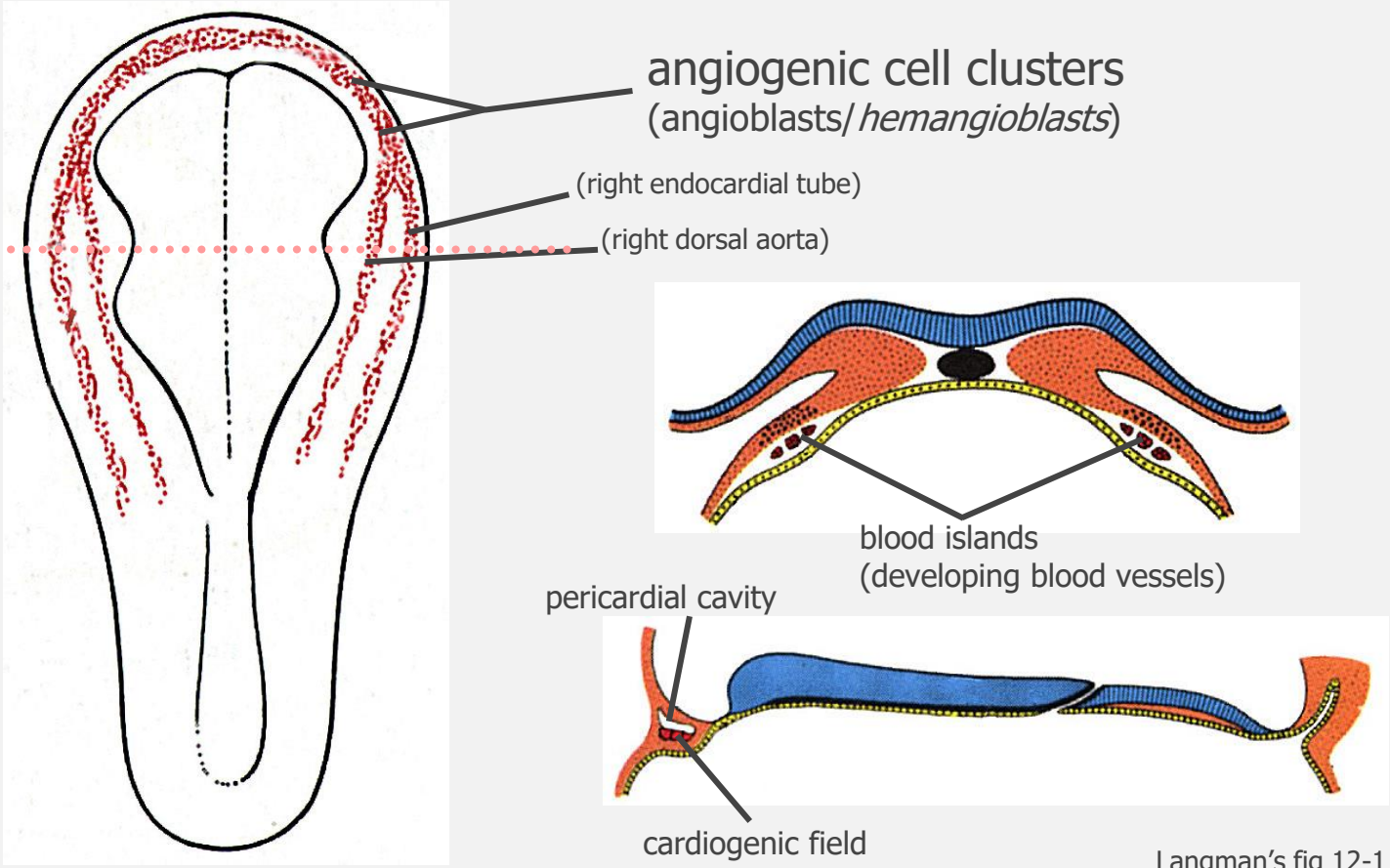
The cardiovascular system is **mesodermally** derived.



Specifically,
lateral splanchnic mesoderm…

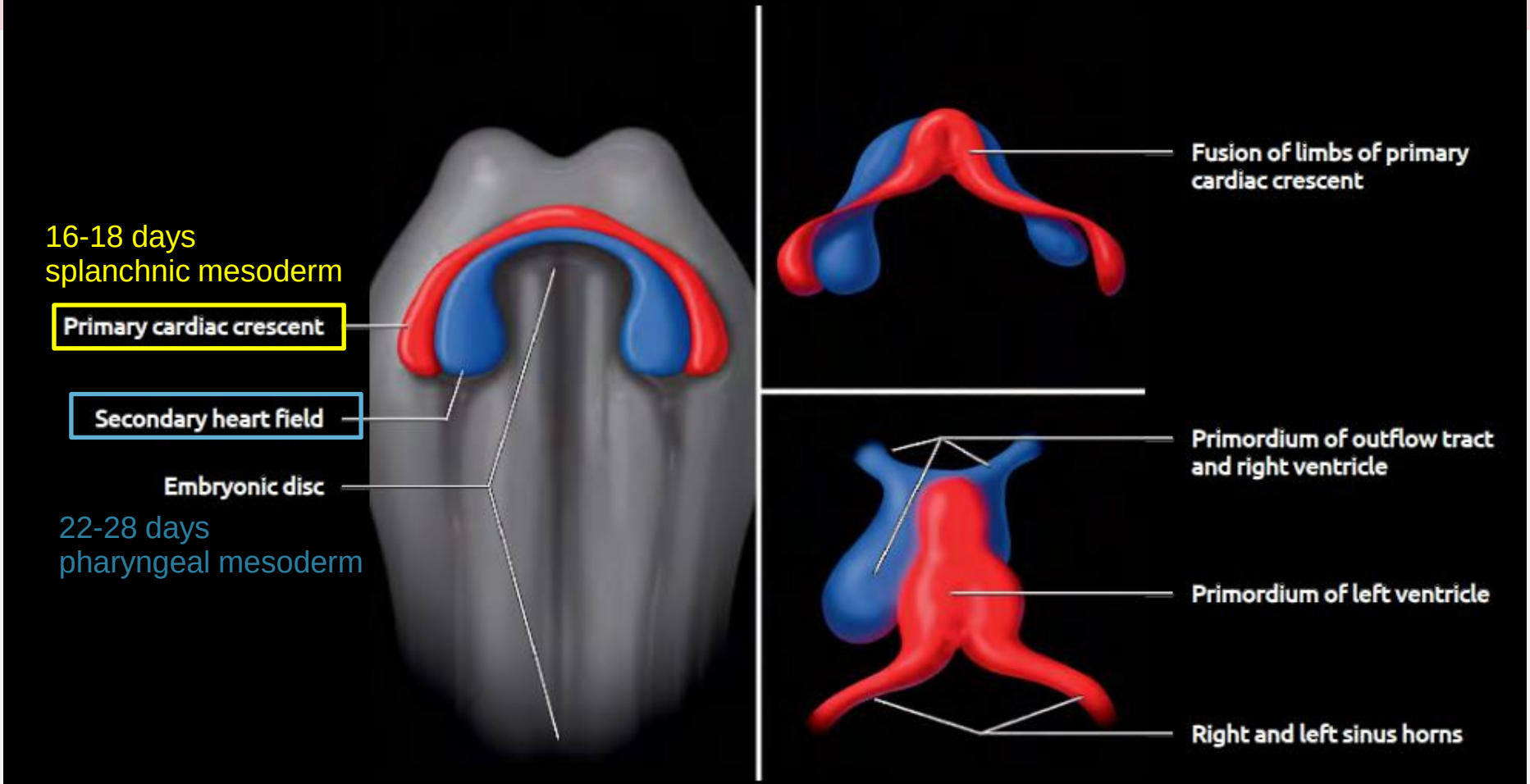
Gilbert fig 14.1

The cardiogenic field is established in the **mesoderm** just after gastrulation (~18-19 days) and develops into a fully functional, multi-chambered heart by the 8th week

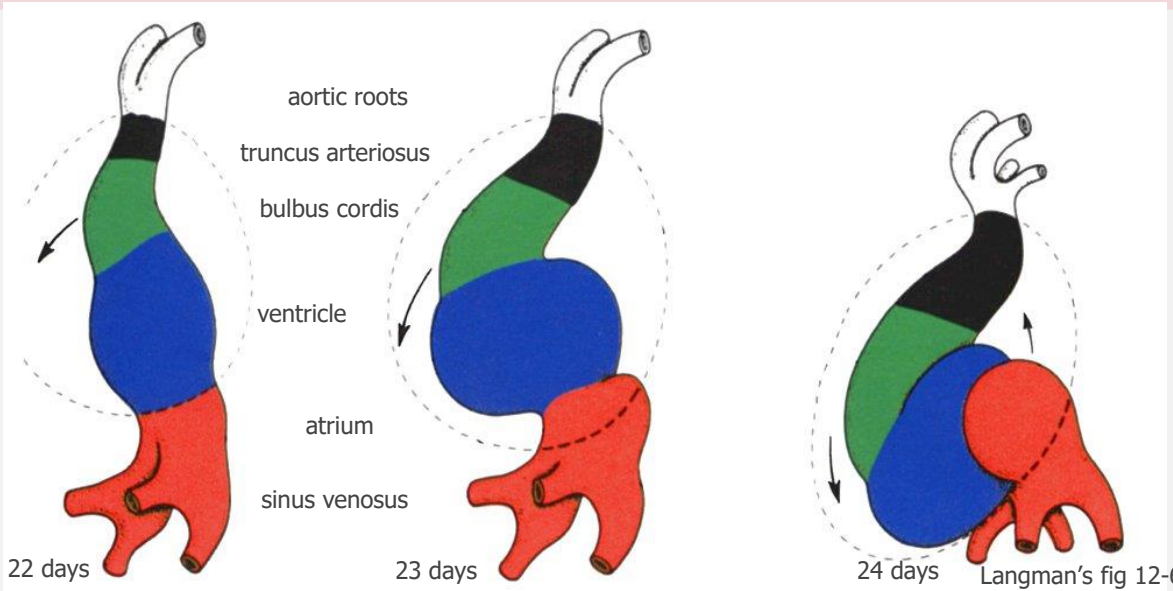


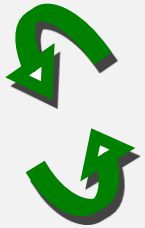
Langman's fig 12-1

PRIMITIVE HEART TUBE

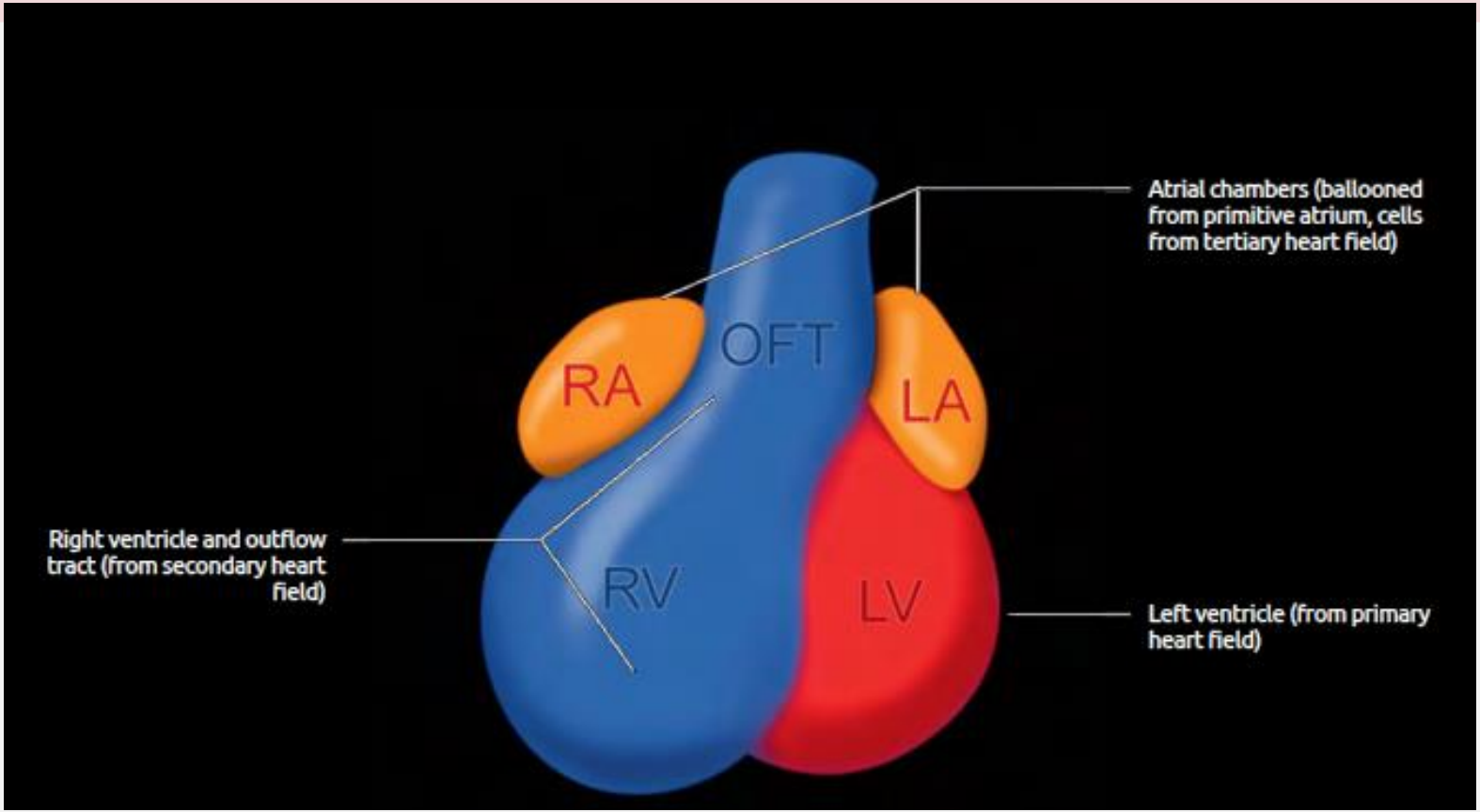


Folding and rotation of heart tube



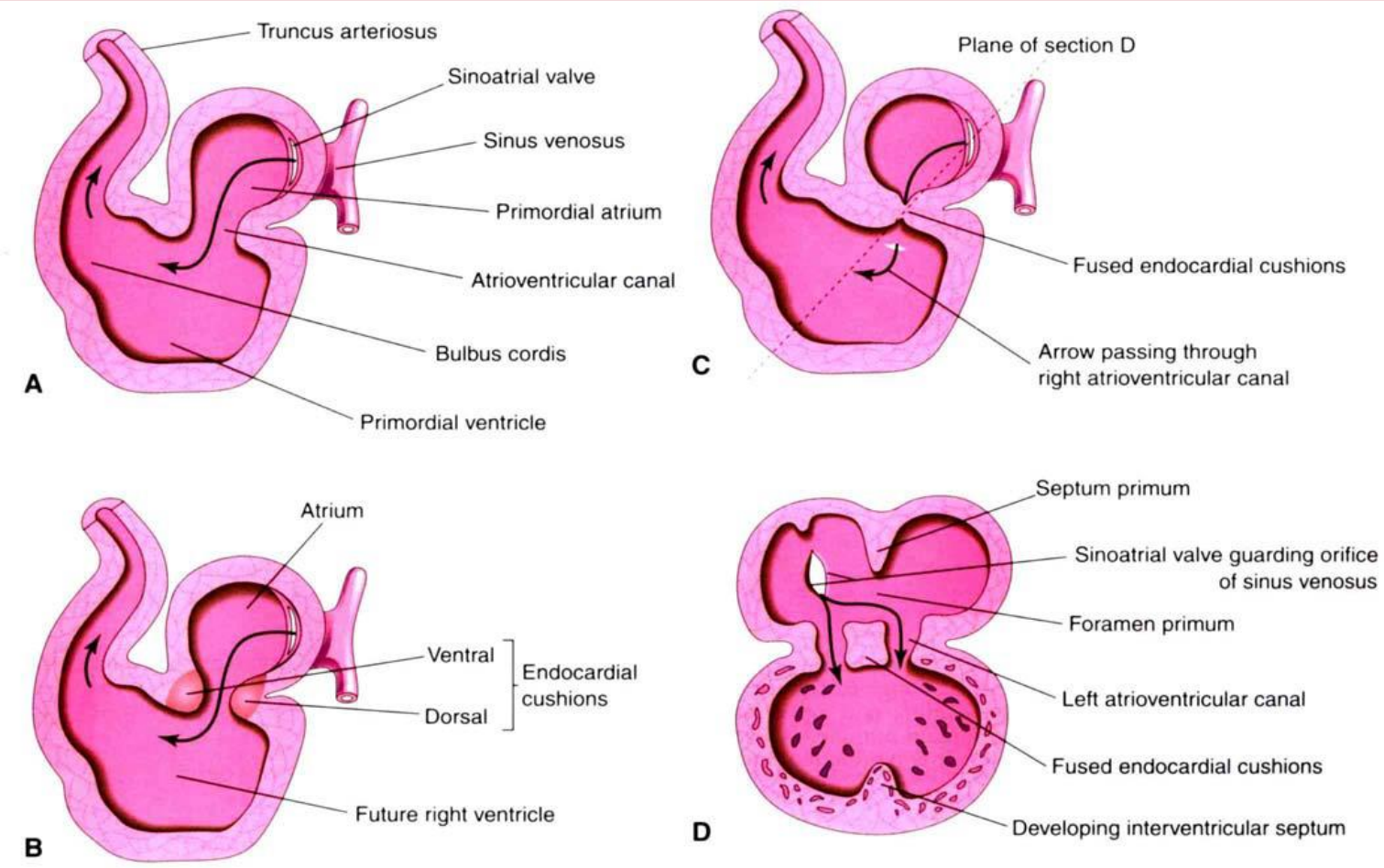


- **Ventricle** moves **ventrally** and to **right**
- **Atrium** moves **dorsally** and to **left**

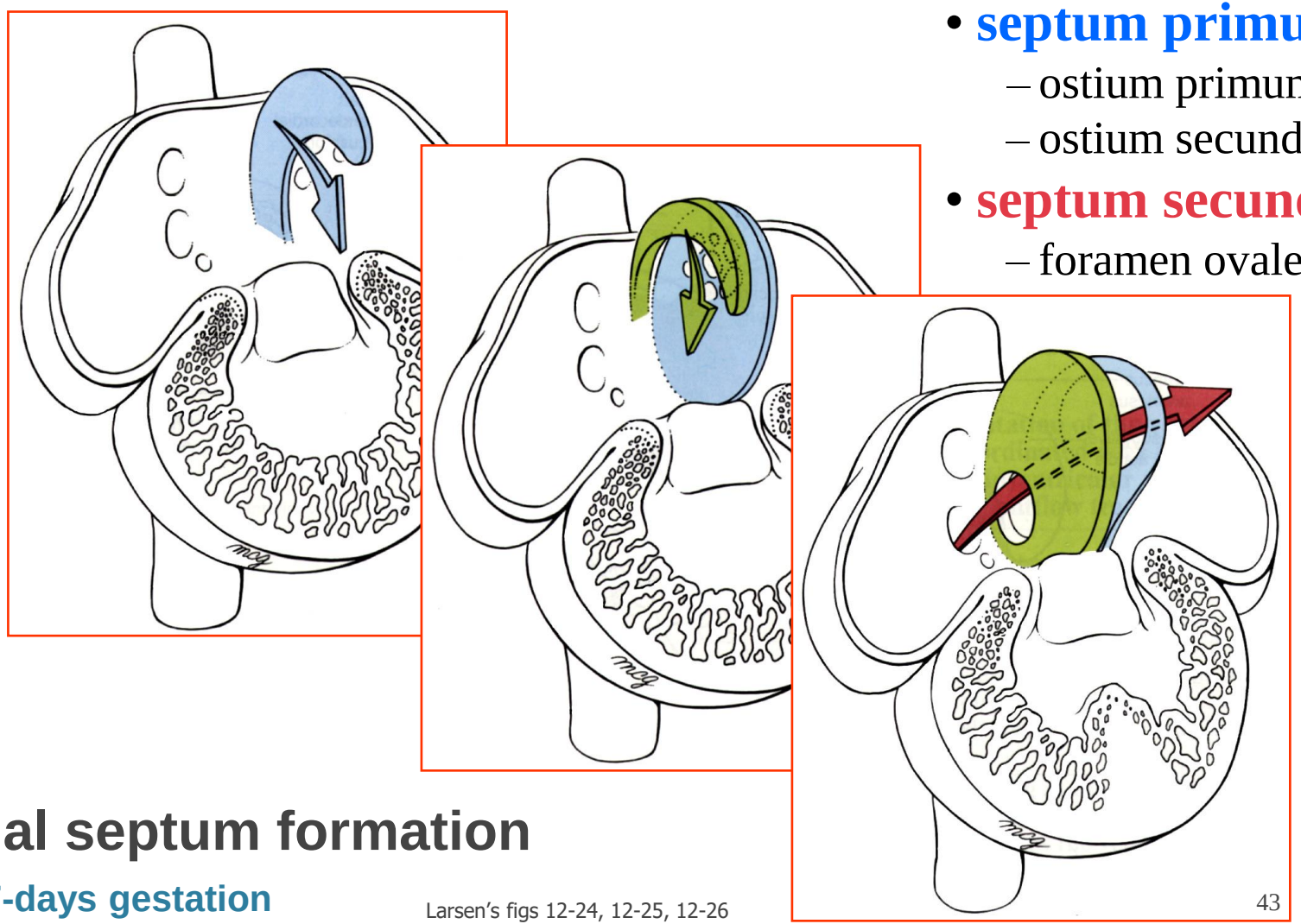

Atrial chambers (ballooned from primitive atrium, cells from tertiary heart field)
RA
OFT
LA
Right ventricle and outflow tract (from secondary heart field)
RV
LV
Left ventricle (from primary heart field)

Atrioventricular Endocardial Cushions

Late 4th weeks



Moore & Persaud fig 13-11



- **septum primum**
  - ostium primum
  - ostium secundum
- **septum secundum**
  - foramen ovale

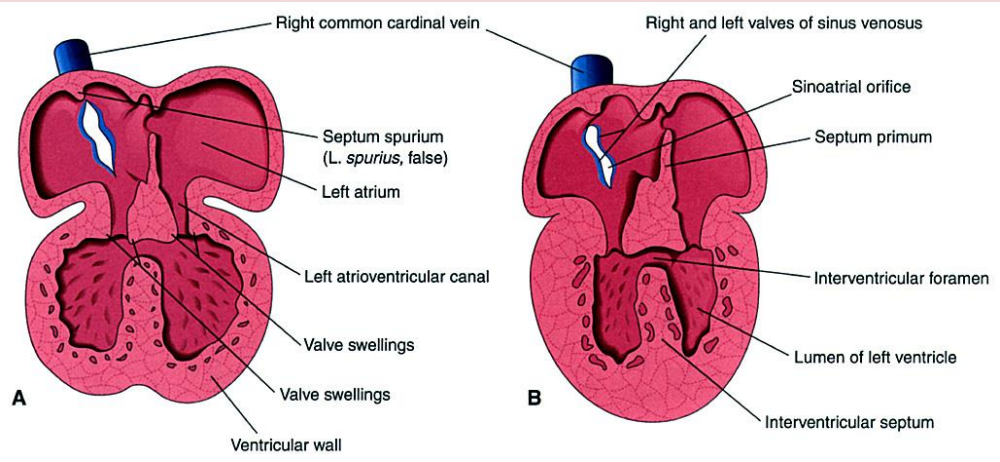
Atrial septum formation

23-27-days gestation

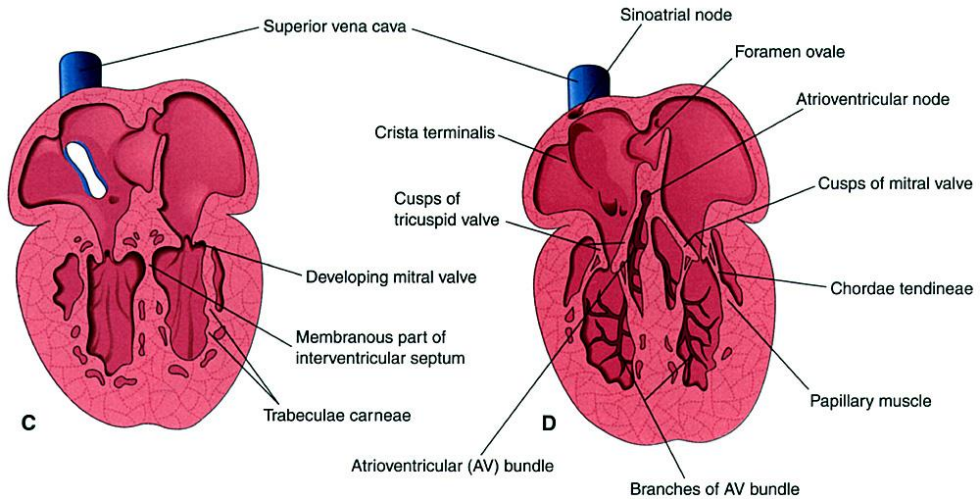
Larsen's figs 12-24, 12-25, 12-26

Formation of AV valves

Late 4th weeks

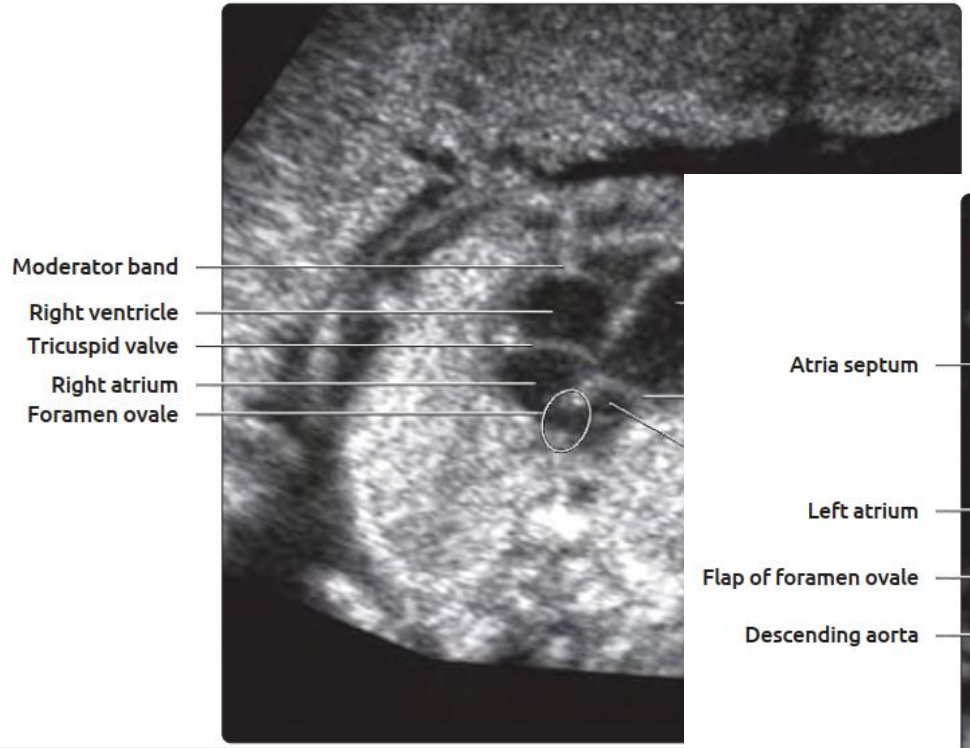


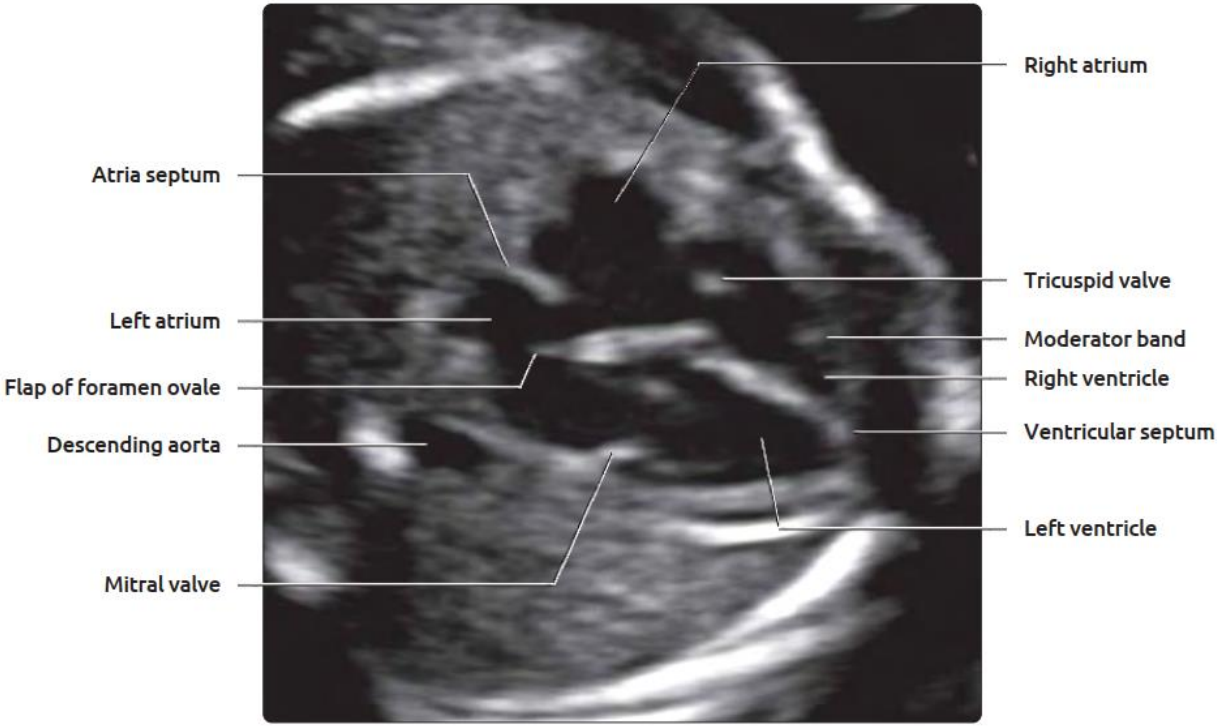
dependent on AV cushions and ventricular myocardium



Moore & Persaud fig 13-19

Four-chamber view





ASD

Incidence and Etiology

- The third most common type of congenital heart disease
- About 10% to 15% of CHD after birth
- Abnormal embryologic development of the atrial septum
- It may be **isolated** or **associated with other cardiac defects** as a part of complex CHD
- Associated with bone malformations and syndromes
  - Holt-Oram, Noonan and Treacher Collins
    → Less frequently
- **Familial** form of **secundum ASD**
  - gene mutations (*GATA4* and *NKX2-5* genes)

Types of ASD

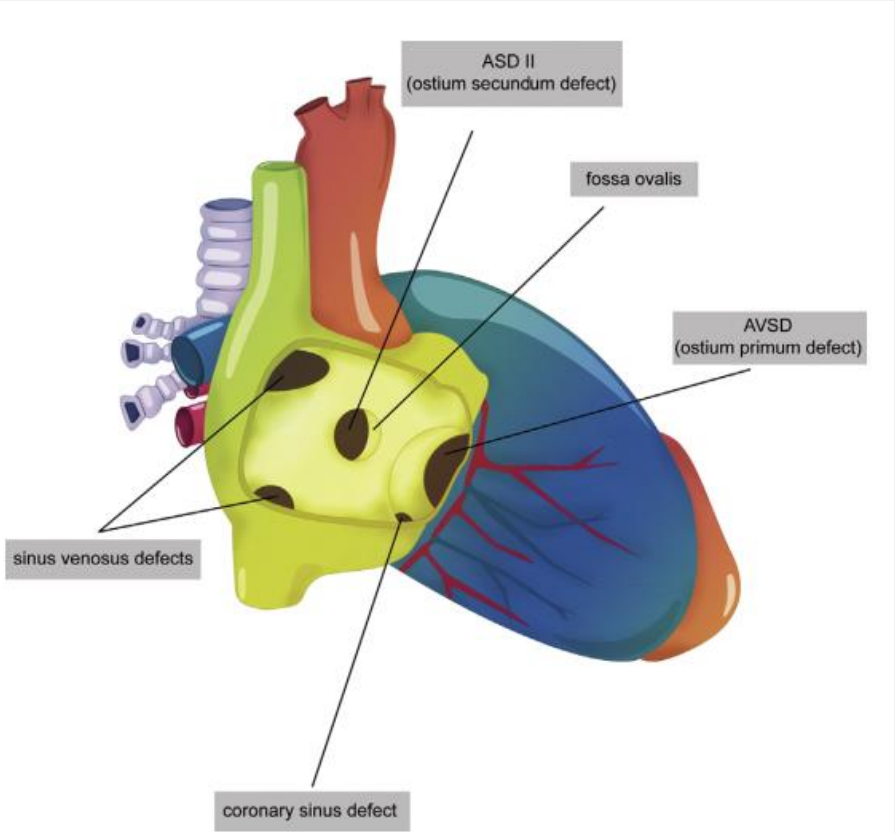
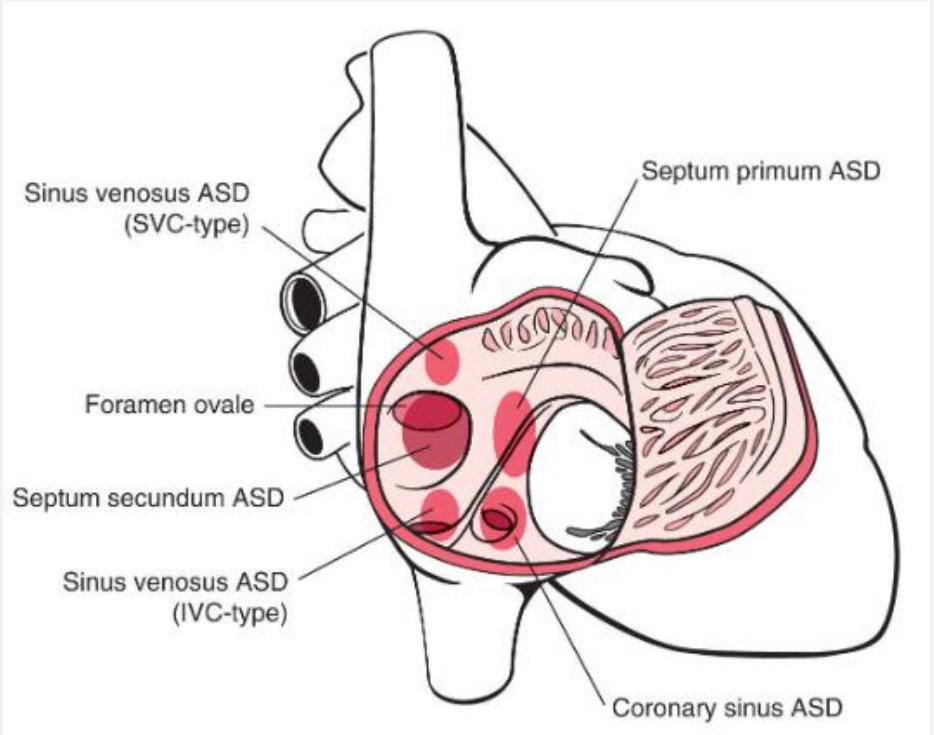


Fig. 9. Types of atrial septal defect (ASD).



N.J. Bravo-valenzuela et al. / Indian Heart Journal 70 (2018) 150–164

Secundum ASD

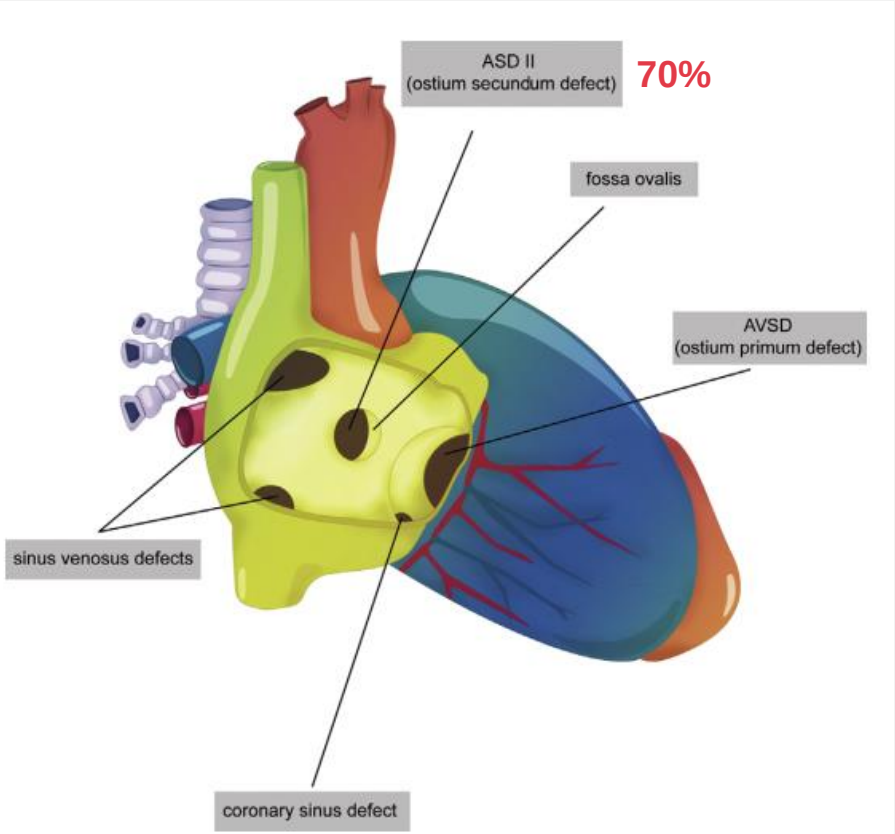
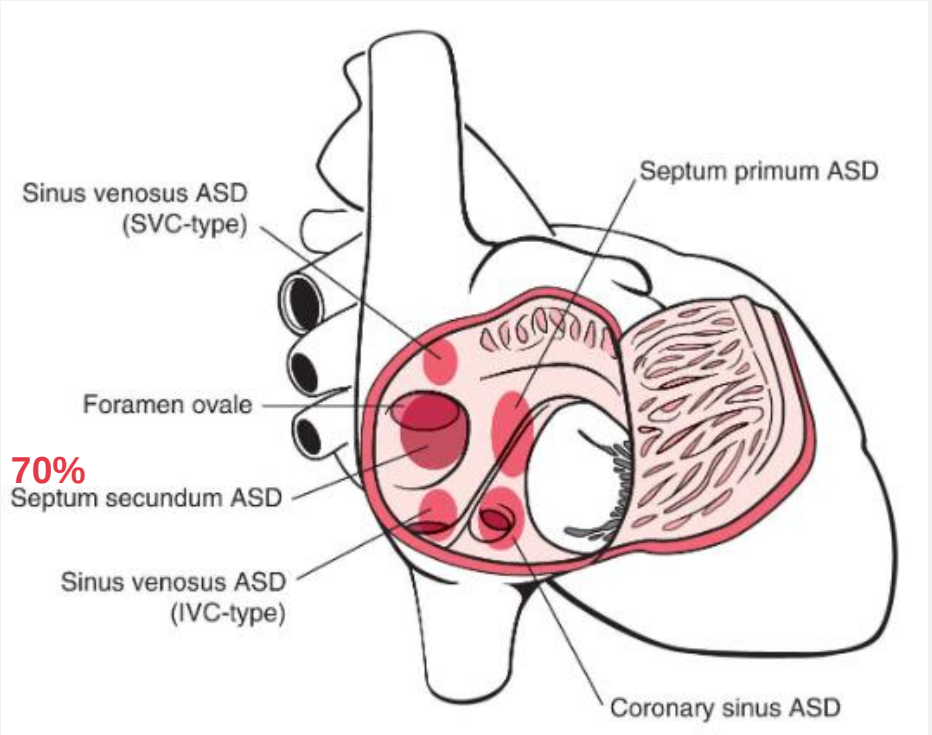
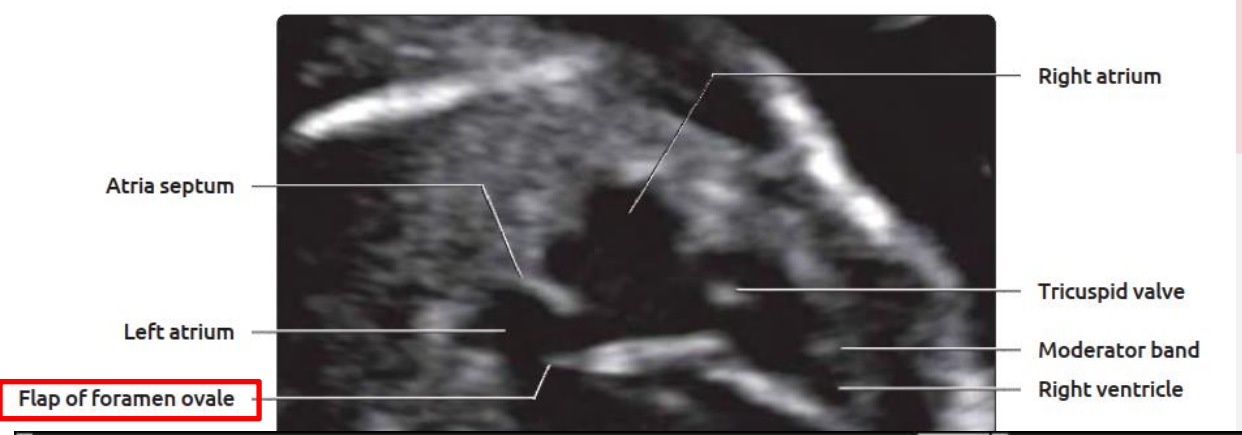


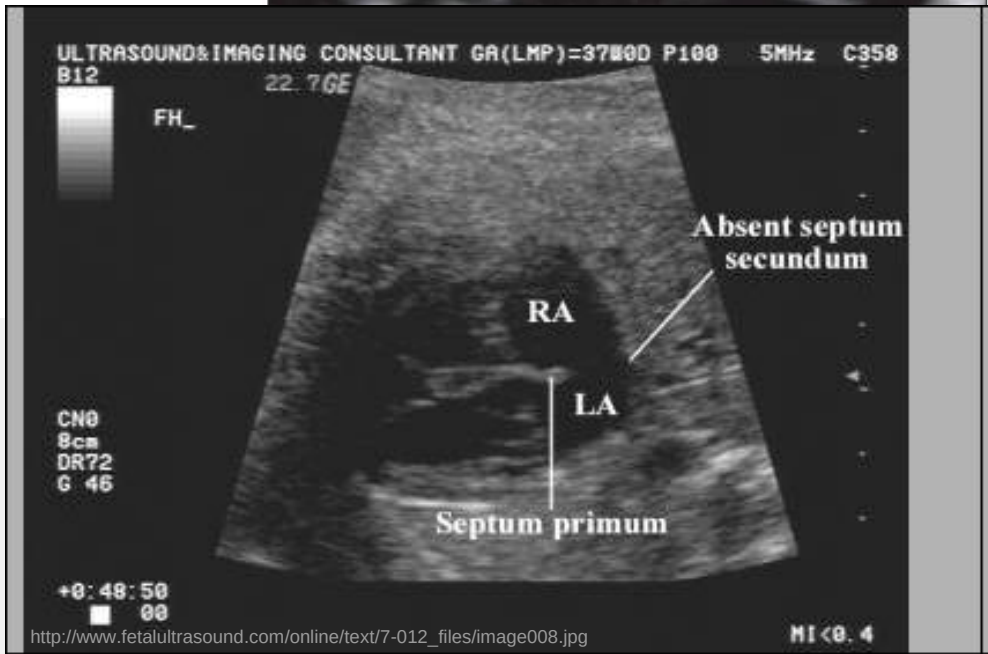
Fig. 9. Types of atrial septal defect (ASD).

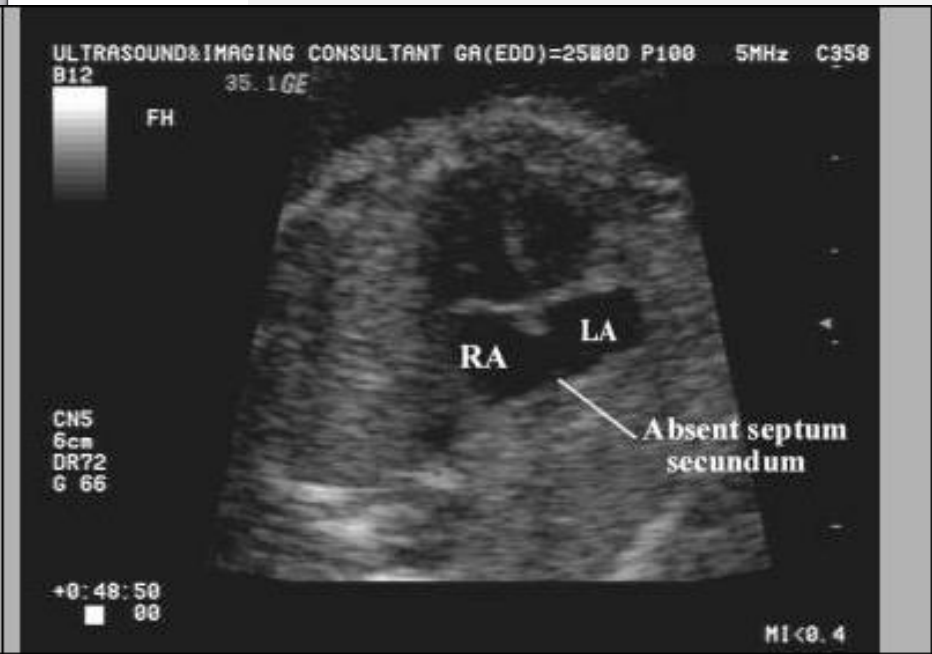


small secundum ASD (<6 mm) close spontaneously

N.J. Bravo-valenzuela et al. / Indian Heart Journal 70 (2018) 150–164

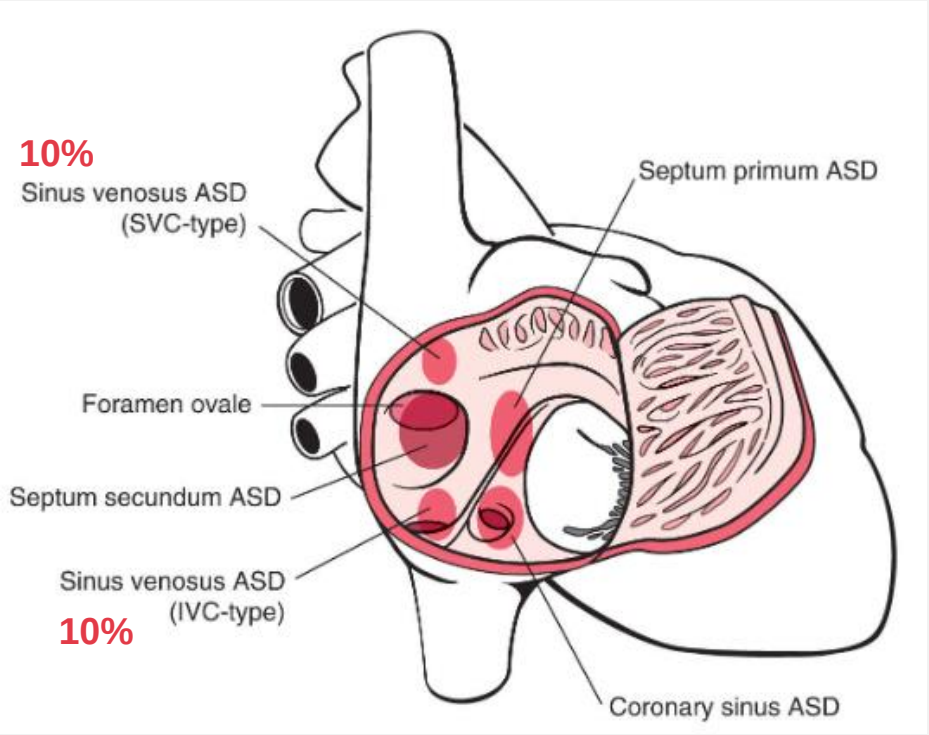




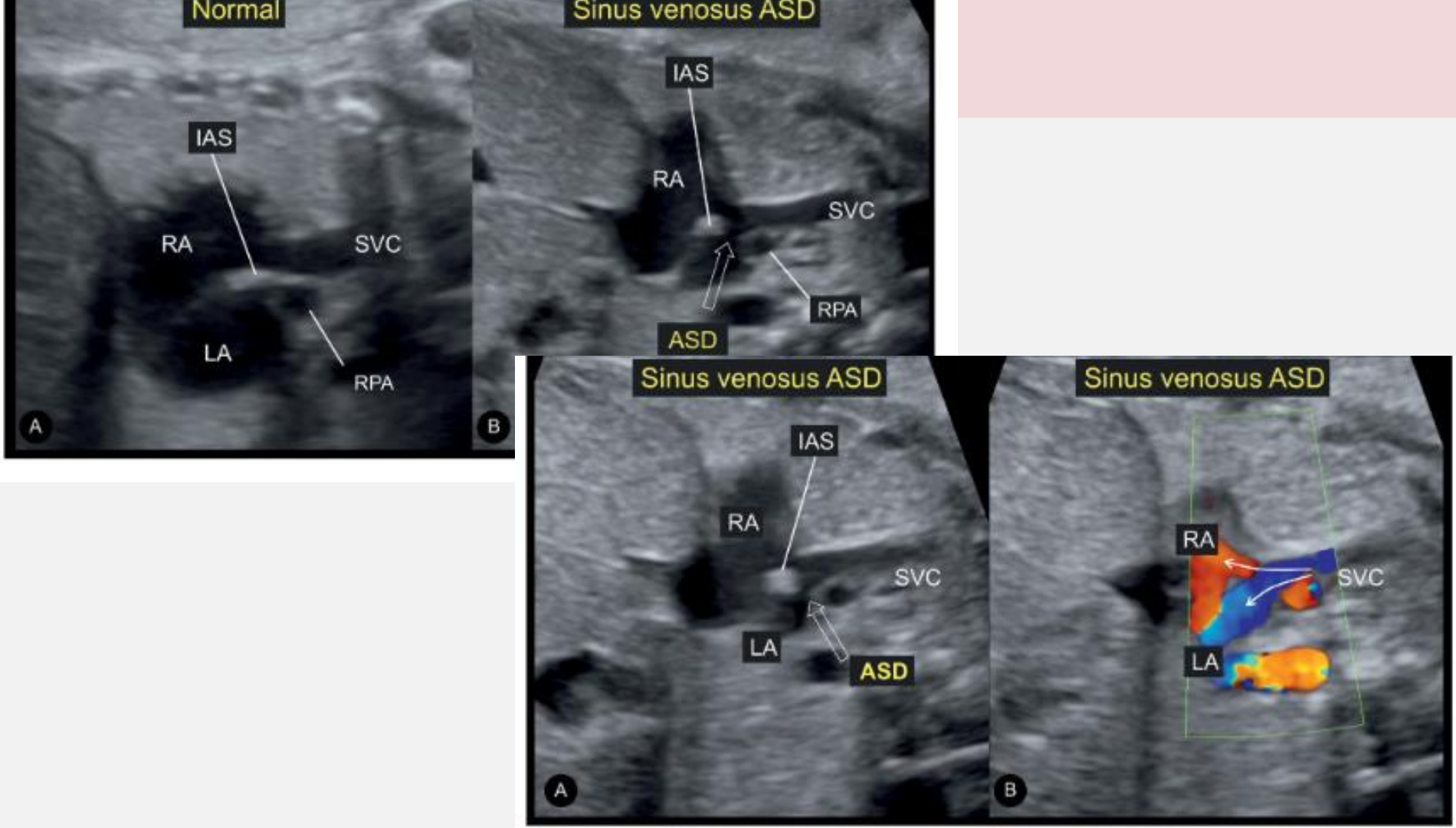


http://www.fetalultrasound.com/online/text/7-012_files/image008.jpg

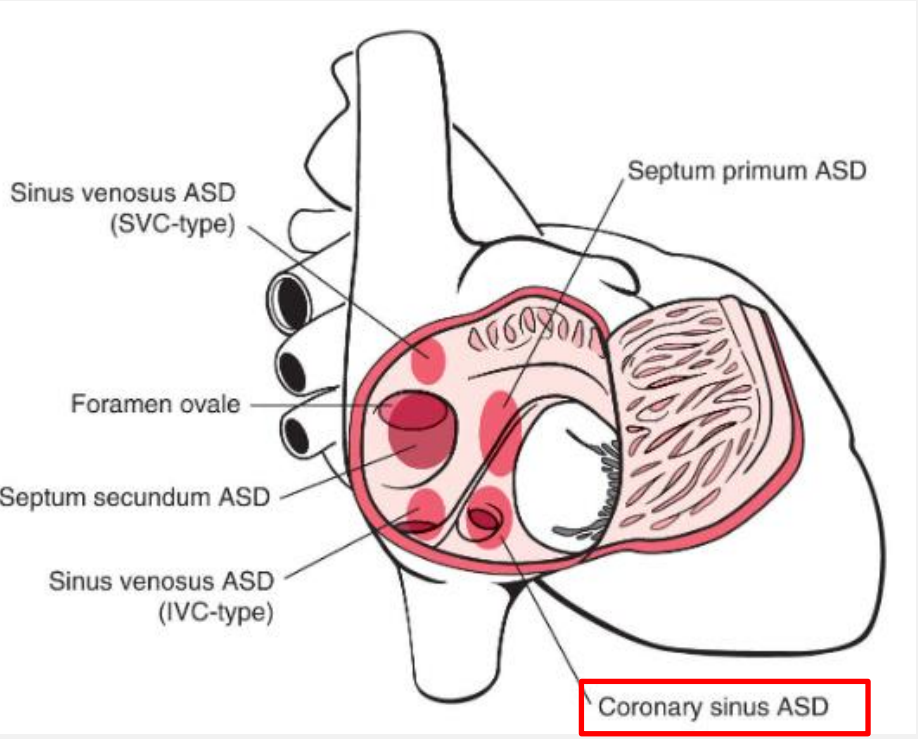
Sinus venous ASD



- A less common septal defect
- Both are frequently associated with anomalous pulmonary venous return.
- The most common location is between the **right upper pulmonary vein** and the **superior vena cava**


Normal
IAS
RA
SVC
LA
RPA
A
Sinus venosus ASD
IAS
RA
SVC
RPA
ASD
B
Sinus venosus ASD
IAS
RA
SVC
LA
ASD
A
Sinus venosus ASD
RA
SVC
LA
B

Coronary sinus ASD



- A rare septal defect
- **Partial or complete unroofing of the tissue** separating the coronary sinus from the LA
- The orifice of the **coronary sinus** in this anomaly is **usually large** as a result of the left-to-right shunt.

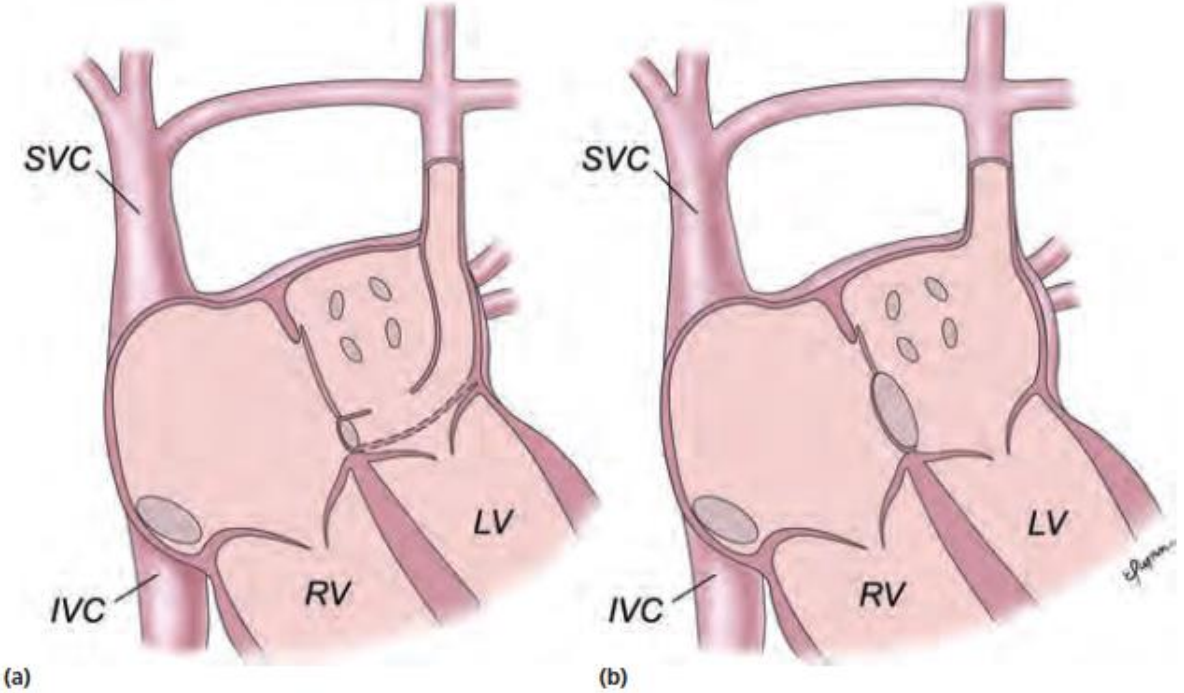


Figure 11.7 Diagram of coronary sinus septal defect. **(a)** Small coronary sinus septal defect associated with left superior vena cava-to-coronary sinus. **(b)** Unroofed coronary sinus associated with left superior vena cava (Raghib syndrome). Note the large interatrial communication through the coronary sinus ostium. IVC, inferior vena cava; LV, left ventricle; RV, right ventricle; SVC, superior vena cava.

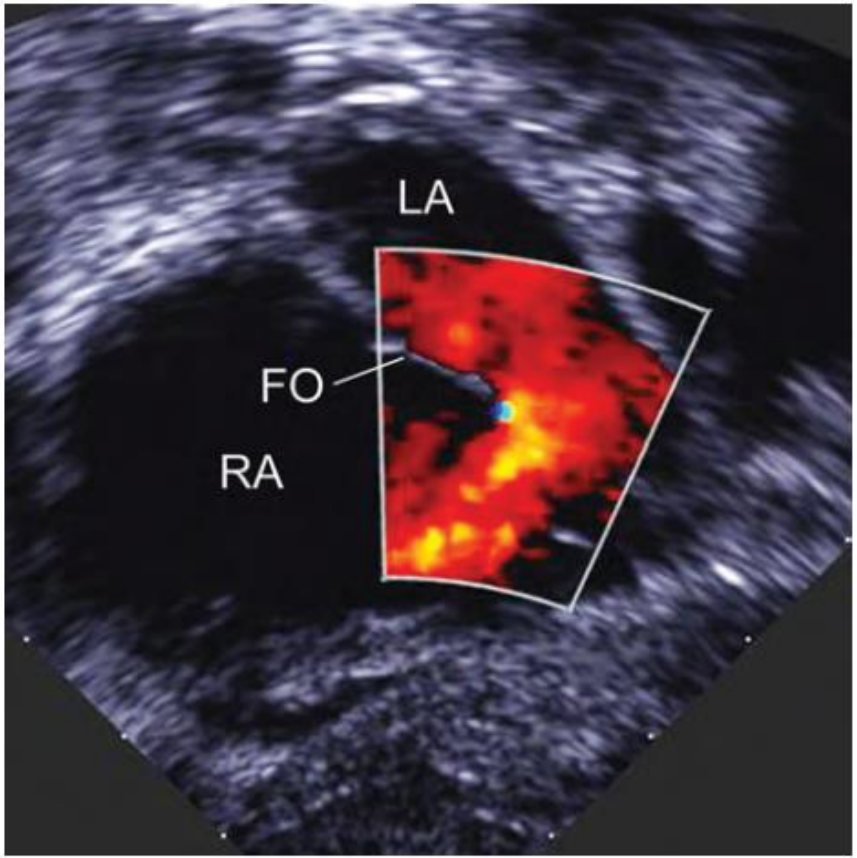


Figure 11.14 Coronary sinus septal defect. Blood flows from the left atrium (LA) through the unroofed coronary sinus into the right atrium (RA) through the enlarged coronary sinus ostium. Note the inferior-posterior location of the coronary sinus ostium and its relation to the intact fossa ovalis (FO).

Primum ASD

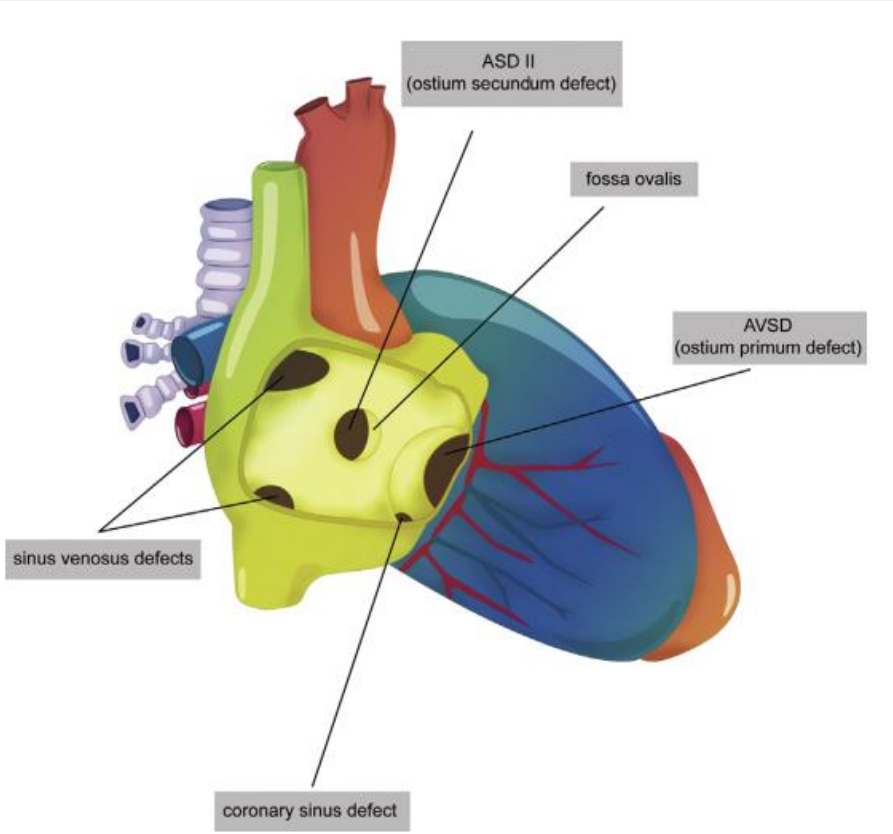
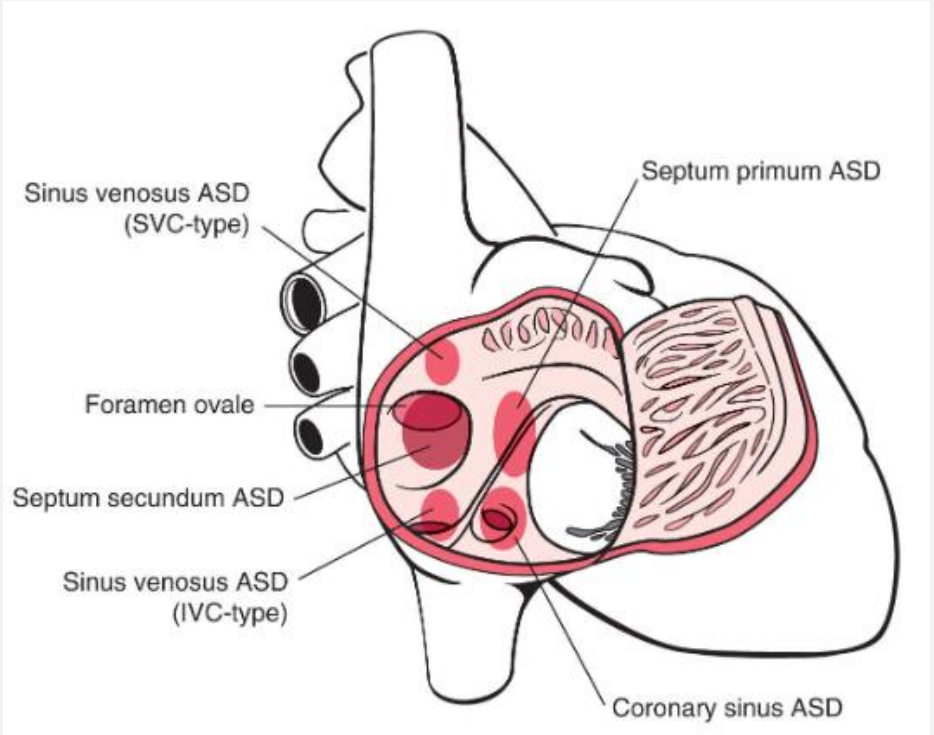
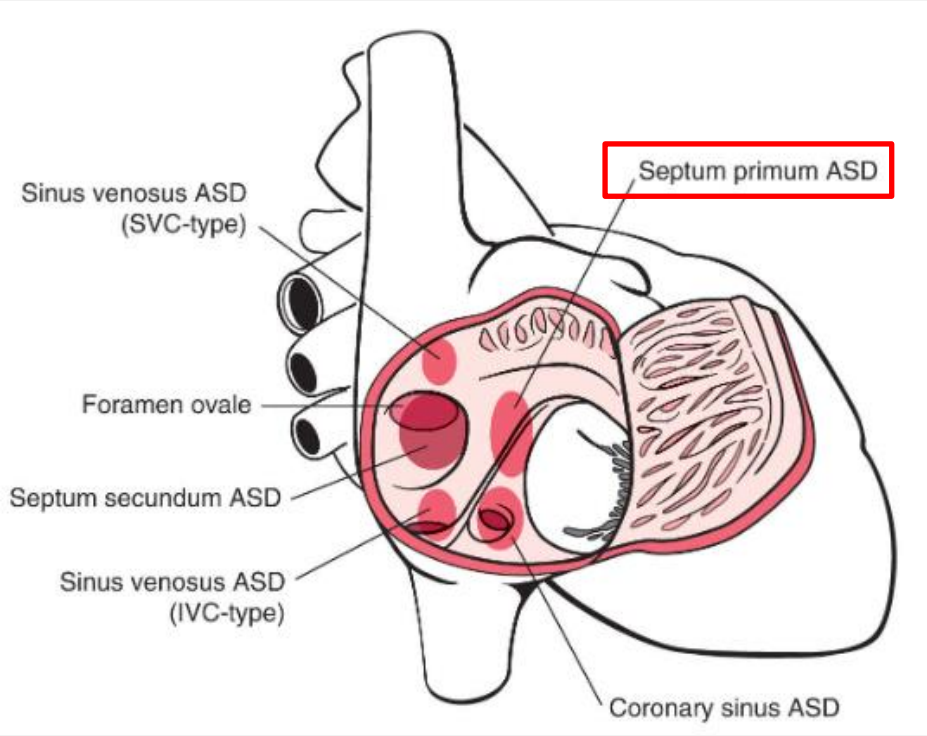


Fig. 9. Types of atrial septal defect (ASD).



N.J. Bravo-valenzuela et al. / Indian Heart Journal 70 (2018) 150–164

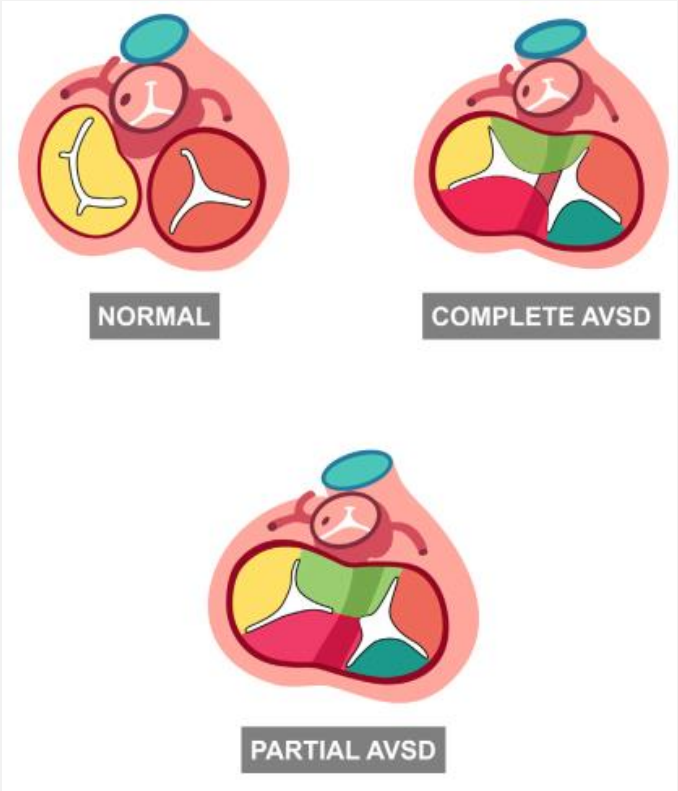
Primum ASD



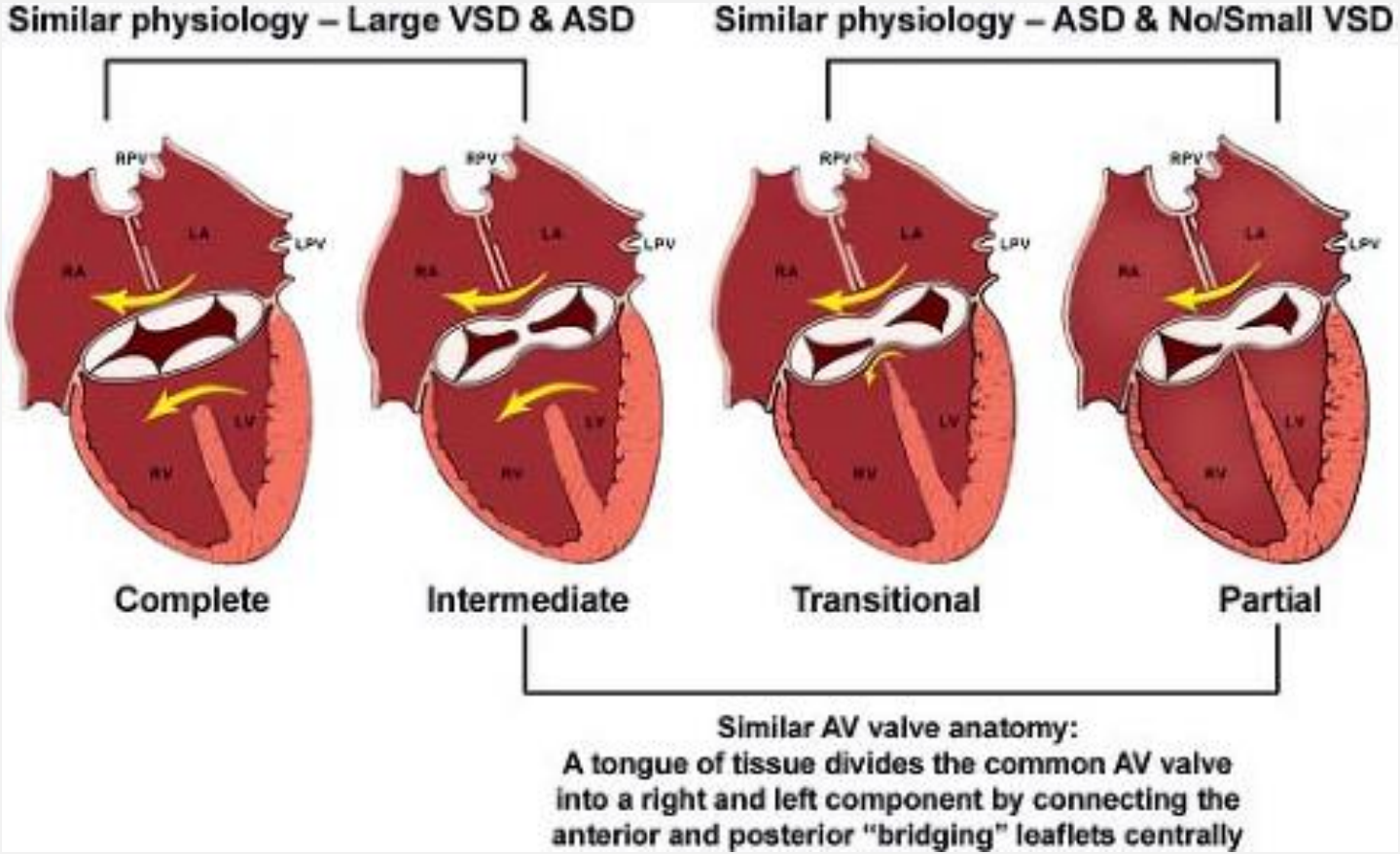
- **Incomplete atrioventricular septal defect**
- An **endocardial cushion defect** with an absent fusion of the lower atrial septum to the underlying atrioventricular valve

AVSD

- **Atrioventricular canal defect** or **endocardial cushion defect**



- **Complete form**
  - Primum ASD, an inlet VSD and a common (single) atrioventricular valve
- **Partial form**
  - Primum ASD, distinct mitral and tricuspid valve annuli, cleft in mitral valve


Similar physiology – Large VSD & ASD
Similar physiology – ASD & No/Small VSD
RPV
LA
LPV
RA
LV
RV
Complete
Intermediate
Transitional
Partial
Similar AV valve anatomy:
A tongue of tissue divides the common AV valve into a right and left component by connecting the anterior and posterior "bridging" leaflets centrally

AVSD

- **Diagnostic pearl of the imaging**
- **Best diagnostic clue**
  - Missing crux of heart on 4-chamber view
    - Normally atrial and ventricular septa meet at crux of heart and AV valves are separated into 2 distinct valve annuli
  - Presence of atrial and ventricular septal defects
  - Usual offset of the AV valves is absent (i.e., valve is in same plane)

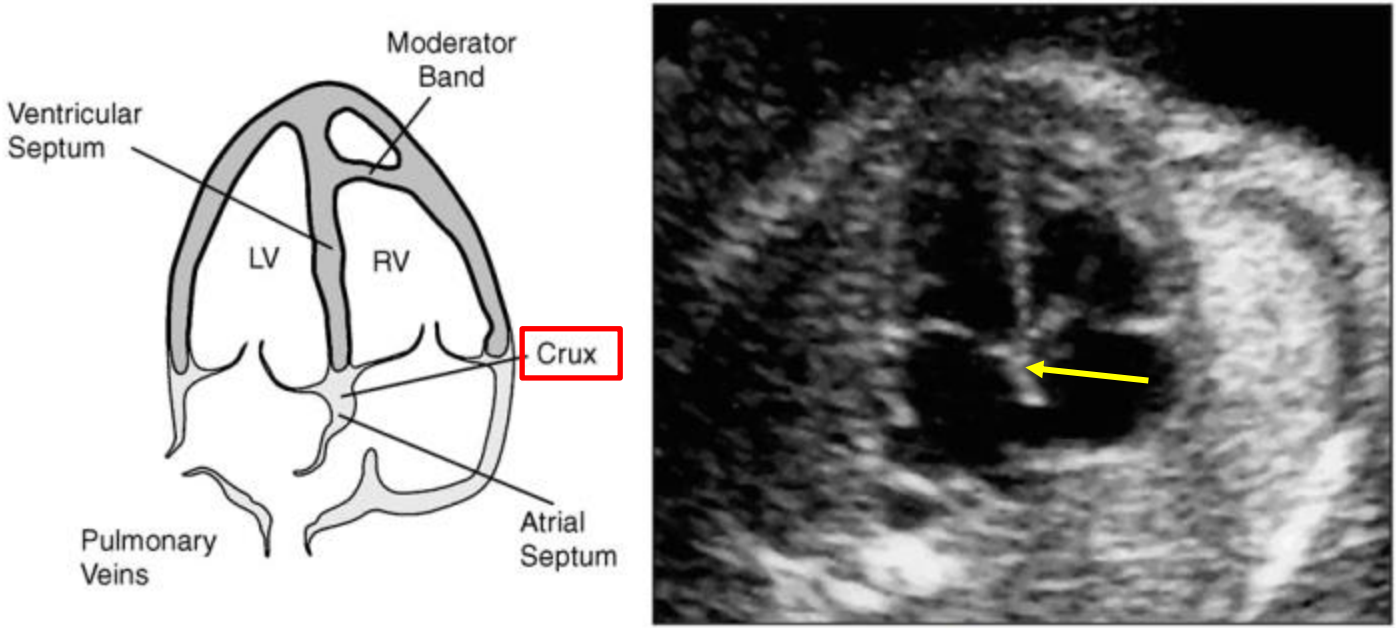
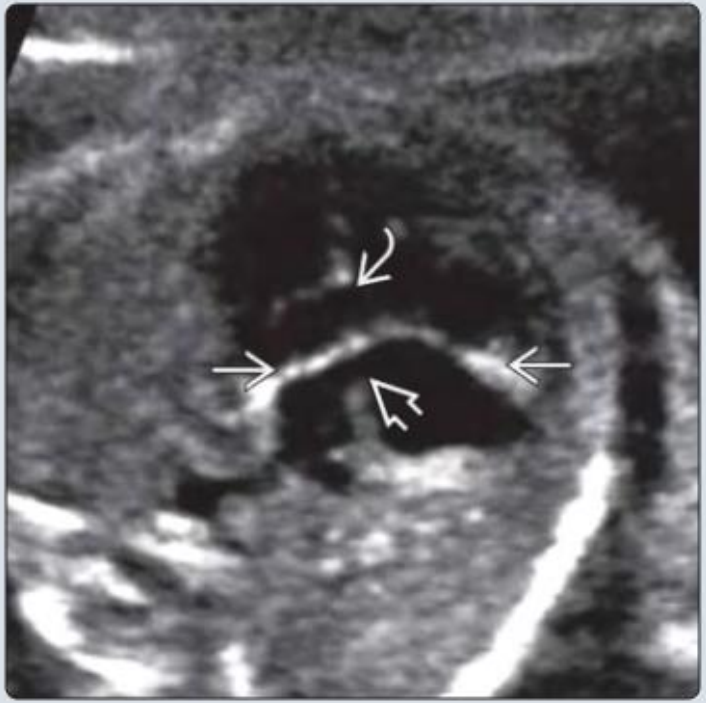
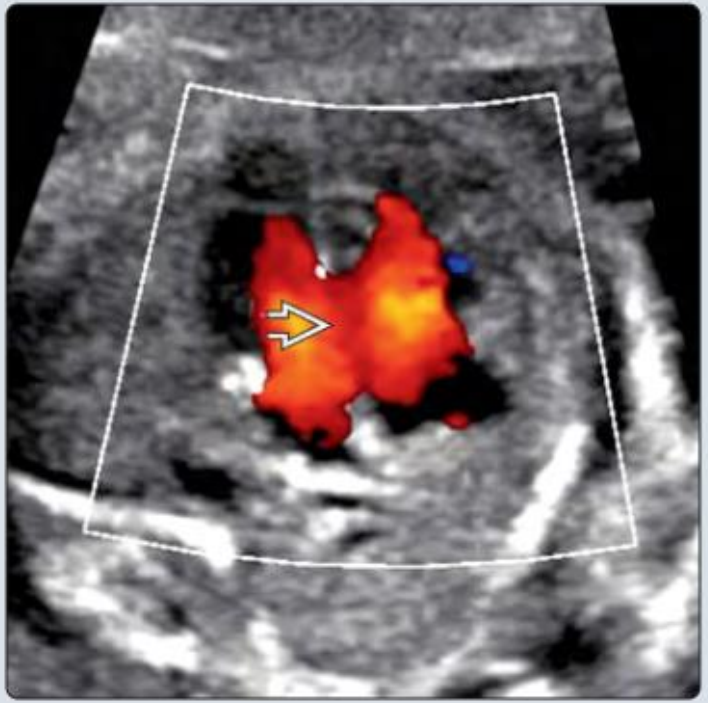


Fig. 2. Four-chamber view of the heart. Key components of a normal four-chamber view include an intact interventricular septum and atrial septum primum. There is no disproportion between the left (LV) and right (RV) ventricles. A moderator band helps to identify the morphologic right ventricle. Note how the "offset" atrioventricular septal valve leaflets insert into the crux. (*From* Lee W. American Institute of Ultrasound in Medicine. Performance of the basic fetal cardiac ultrasound examination. J Ultrasound Med 1998;17:601–7; with permission.)

(Left) *Four-chamber view fetal echocardiogram shows a balanced atrioventricular septal defect (AVSD) with a single common AV valve ⇨ in systole and contiguous atrial ⇨ and ventricular ⇨ septal defects. The usual offset of the valves is absent.* **(Right)** *Color Doppler image in diastole shows blood filling the entire atrioventricular septal defect ⇨. The crux of the heart is missing and there is complete mixing of oxygenated and deoxygenated blood.*





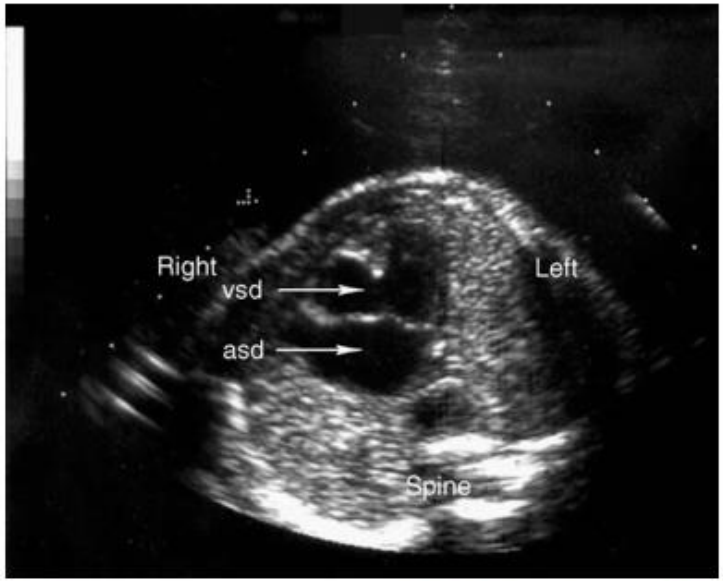


Fig. 12. Atrioventricular septal defect. Sonographic findings for a complete atrioventricular septal defect consist of atrial septal defect (asd), ventricular septal defect (vsd), and lack of the normal offset atrioventricular valve insertion sites. The common valve appears as a straight echogenic line.

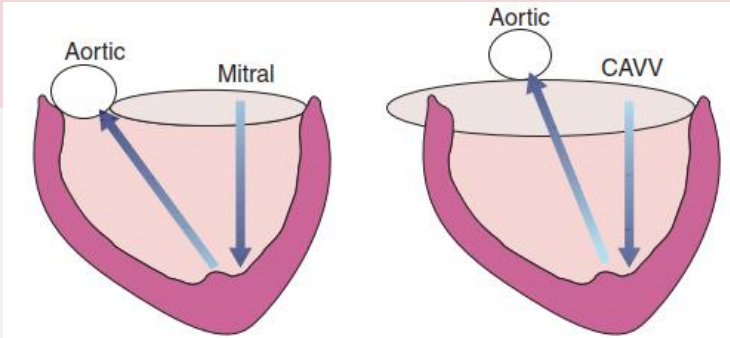


Figure 15.5 Diagram showing that the length of inflow and outflow are the same in the left ventricle of the normal heart. In CAVC, the outflow length is elongated because of the unwedged aortic valve.

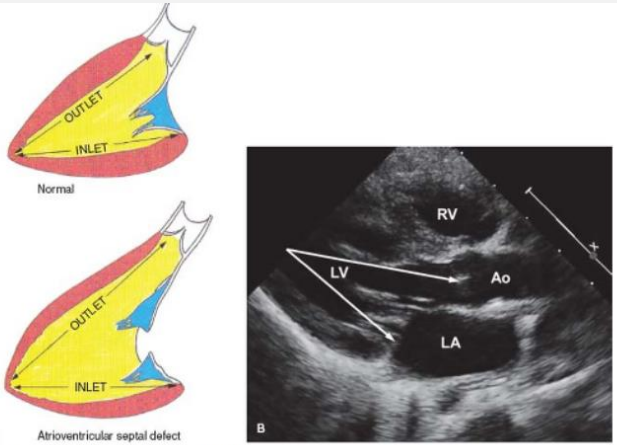
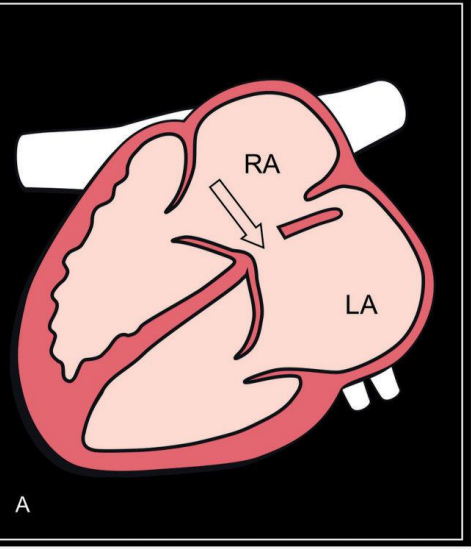
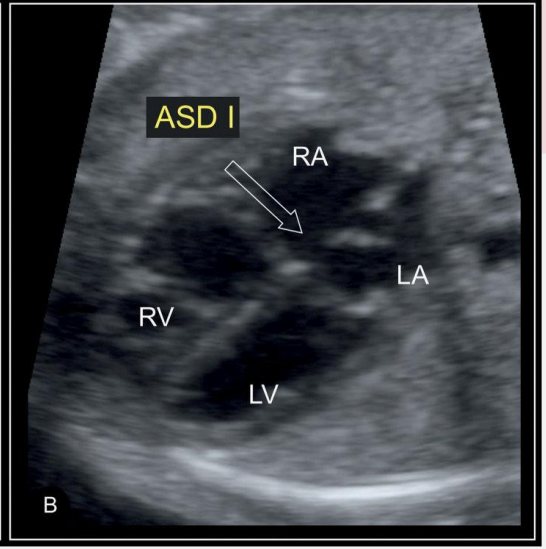
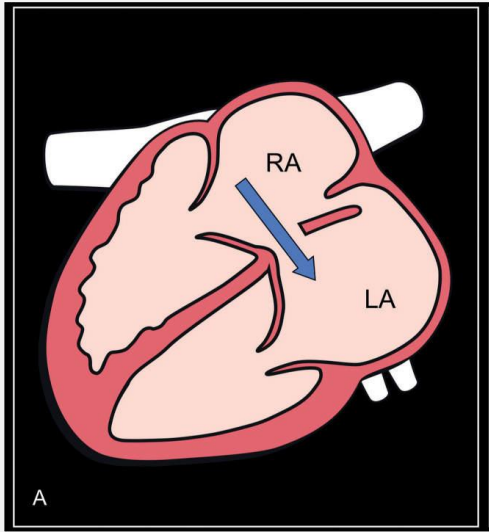
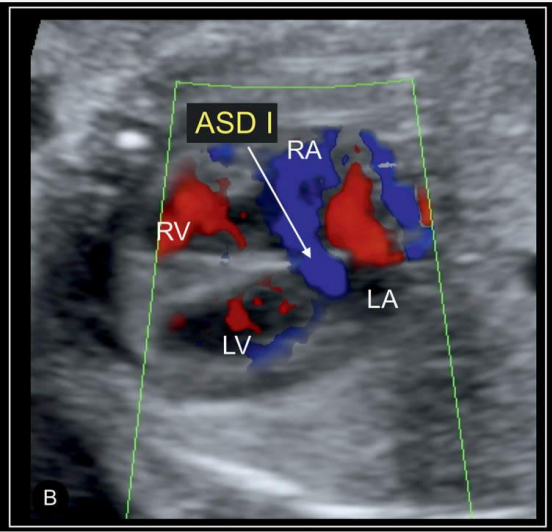


Figure 29.3 Elongated left ventricular outflow tract (LVOT) in atrioventricular septal defect (AVSD). Demonstrated in a diagram (**A**) and parasternal long-axis echocardiograph (**B**). Because of deficiency of the ventricular component of the atrioventricular septum and the "sprung" atrioventricular junction, the distance from the LV apex to the posterior left atrioventricular valve annulus is 20% to 25% shorter than the distance from the apex to the aortic annulus. Ao, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle. (A courtesy of Robert H. Anderson, MD.)


RA
LA
A


ASD I
RA
LA
RV
LV
B


RA
LA
A


ASD I
RA
RV
LA
LV
B

AVSD

- **Complete AVSD** is associated with extra-cardiac malformations and syndromes such as **trisomies 21 (75% of cases), 18 and 13**.
- Genetics
  - Trisomy 21 in up to 50% of fetal cases
  - Other chromosomal anomalies or syndromes in 20-30%
    - Trisomy 18, 13, heterotaxy syndromes
- Associated abnormalities
  - Heterotaxy found in 15-20%
  - Additional cardiac malformations, such as tetralogy of Fallot, double outlet right ventricle, left heart obstruction
    - Found in 10% with trisomy 21
    - Found in 33% in non-Down syndrome group

AVSD

- **Imaging Recommendations**
  - Monitor for signs of hydrops
    - Pericardial effusion, pleural effusion, ascites, skin edema
    - Cardiomegaly
      - Track ratio of heart to chest circumference
  - Full anatomic survey for other anomalies
    - Strong association with trisomy 21
      - Thickened nuchal fold, rhizomelic limb shortening, duodenal atresia, echogenic bowel, pelviectasis, clinodactyly

VSD

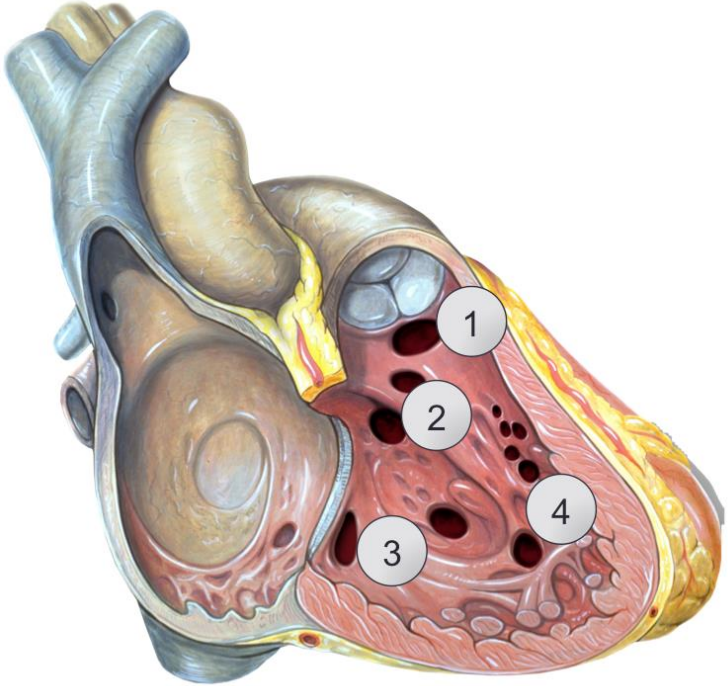
Incidence and Etiology

- The most common CHD occurring in about 20-30% of neonates with CHD
- Can occur **sporadically** or in association with **gene mutations**
  - *TBX5* and *GATA4* genes
- Most of all, VSDs evolve to spontaneous closure, even in utero or during the first year of life.
- Associated cardiac abnormalities present in 50%

VSD

Classification of VSD

- **Type 1**
  - **5% to 7%** of isolated VSD
  - "Conal", "sub-pulmonary", "infundibular", "supracristal"
- **Type 2**
  - **70% of VSDs**
  - "Peri-membranous", "para-membranous", "conoventricular"
- **Type 3**
  - **5% of VSDs, typically occurs in Down syndrome**
  - "Peri-inlet", "AV canal", "AV septal", "endocardial cushion"
- **Type 4**
  - **20% of VSDs**
  - Muscular defect



Source: BMJ Group Best Practice. **Ventricular septal defects**. Illustration by Patrick J. Lynch, medical illustrator

VSD

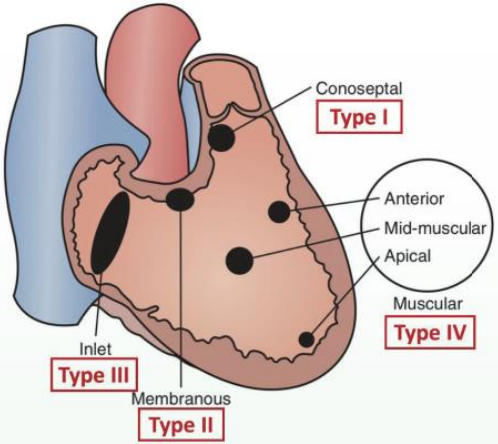
Diagnostic pearl of the imaging

- Keep sound beam **perpendicular** **to septum**
  - Avoids VSD mimic of dropout at membranous-muscular junction

- Look for **septal continuity** with aortic annulus in left ventricular outflow tract (LVOT) view to **exclude membranous VSD**

- The evaluation of LVOT (five-chamber view) helps to identify **outlet** defects, mainly membranous outlet VSD.

圖二：從右心室綜觀心室中膈缺損四種類型



圖片引用至 Callen's Ultrasonography in Obstetrics and Gynecology-6th edition, Figure.13-43

圖三：心室中膈缺損 Type I

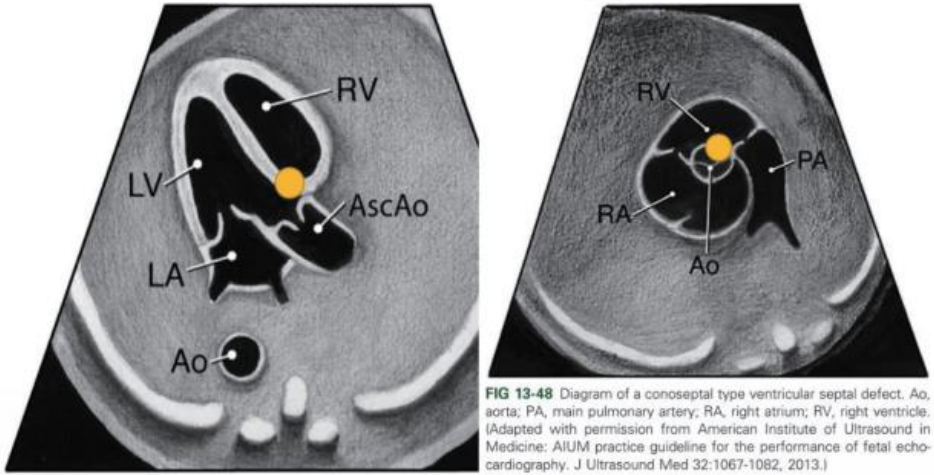


FIG 13-48 Diagram of a conoseptal type ventricular septal defect. Ao, aorta; PA, main pulmonary artery; RA, right atrium; RV, right ventricle. (Adapted with permission from American Institute of Ultrasound in Medicine: AIUM practice guideline for the performance of fetal echocardiography. J Ultrasound Med 32:1067-1082, 2013.)

圖片引用至 Callen's Ultrasonography in Obstetrics and Gynecology-6th edition, Figure.13-48

圖四：心室中膈缺損 Type II

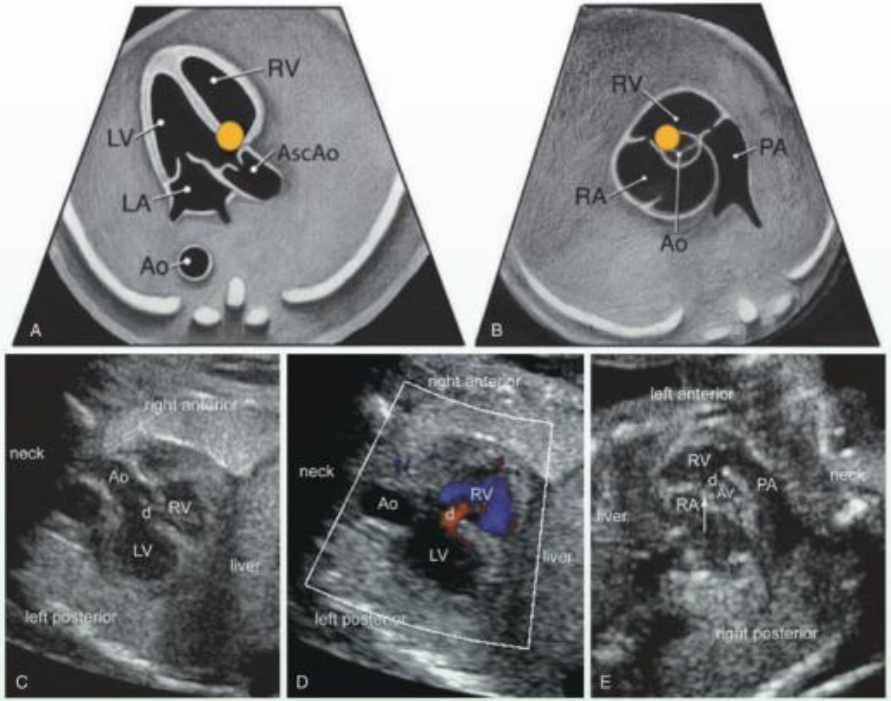
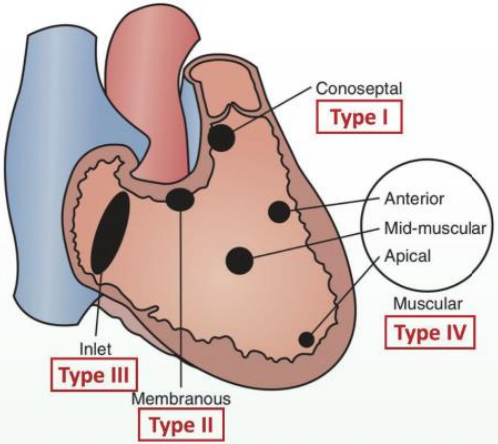


FIG 13-44 Diagrams and sonograms of membranous (also known as perimembranous) ventricular septal defects by two-dimensional and color Doppler imaging. A membranous defect is typically bordered by the tricuspid valve and the aortic valve and is best seen in a left ventricular outflow tract view or a high short-axis view, as shown by the asterisks in **A** and **B**, and the letter "d" in **C** through **E**. There is remnant outlet septum seen in **E** (*asterisk*) anterosuperior to the defect. By color Doppler (**D**), shunting across the defect is typically bidirectional; in this image there is left-to-right shunting in a systole frame. Ao, aorta; AscAo, ascending aorta; AV, aortic valve; d, ventricular septal defect; LA, left atrium; LV, left ventricle; PA, main pulmonary artery; RA, right atrium; RV, right ventricle.

圖片引用至 Callen's Ultrasonography in Obstetrics and Gynecology-6th edition, Figure.13-44

圖二：從右心室縱觀心室中膈缺損四種類型



圖片引用至 Callen's Ultrasonography in Obstetrics and Gynecology-6th edition, Figure.13-43

圖五：心室中膈缺損 Type III

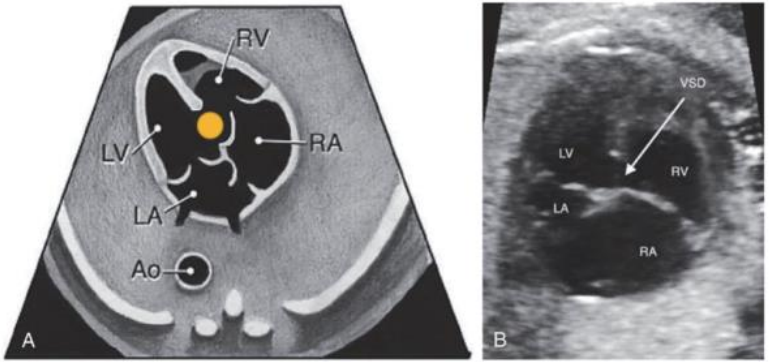


FIG 13-47 Diagram **(A)** and sonographic image **(B)** of atrioventricular canal type (also known as perimembranous inlet) ventricular septal defects (VSDs). VSDs are indicated by an asterisk in A, and the arrow in B. Note that there are two atrioventricular valves, and there is no primum atrial septal defect in this defect. Ao, aorta; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. (**A,** Adapted with permission from American Institute of Ultrasound in Medicine: AIUM practice guideline for the performance of fetal echocardiography. J Ultrasound Med 32:1067-1082. 2013.)

圖片引用至 Callen's Ultrasonography in Obstetrics and Gynecology-6th edition, Figure.13-47

圖六：心室中膈缺損 Type IV

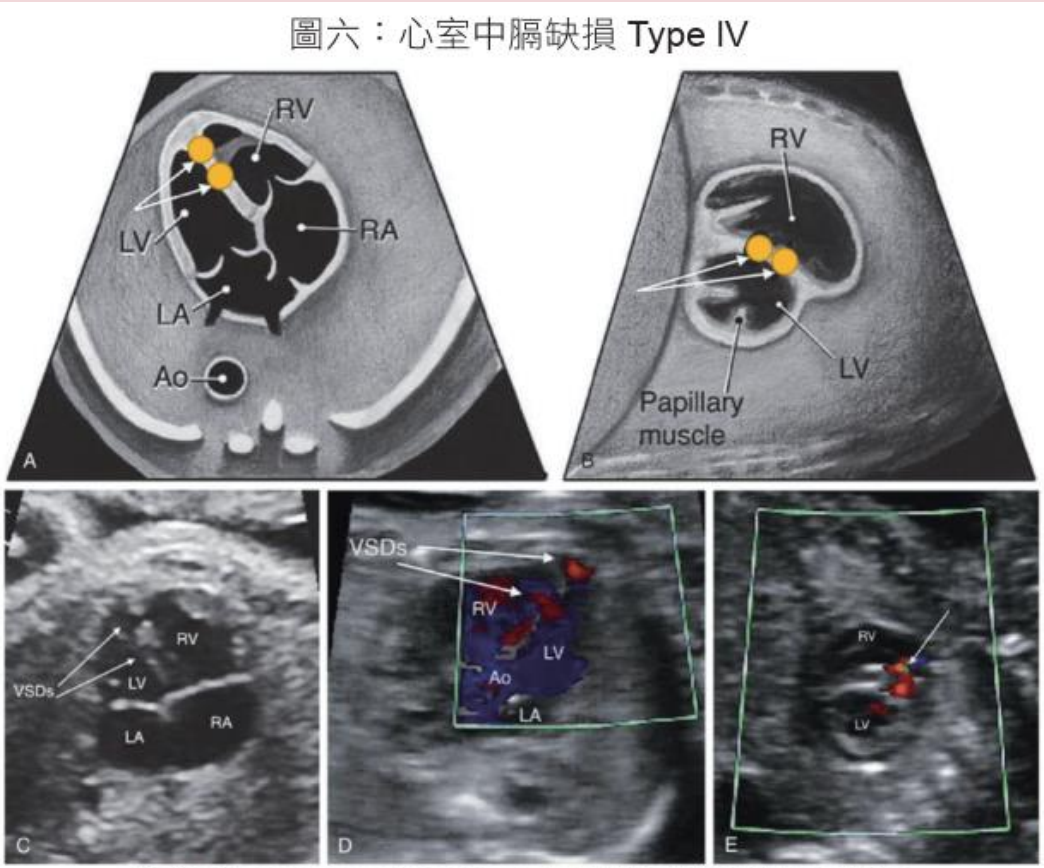
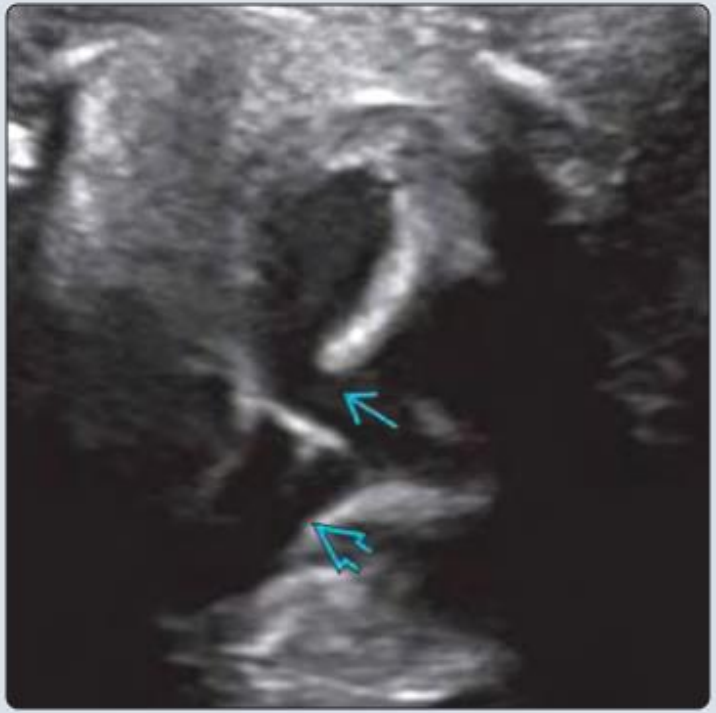
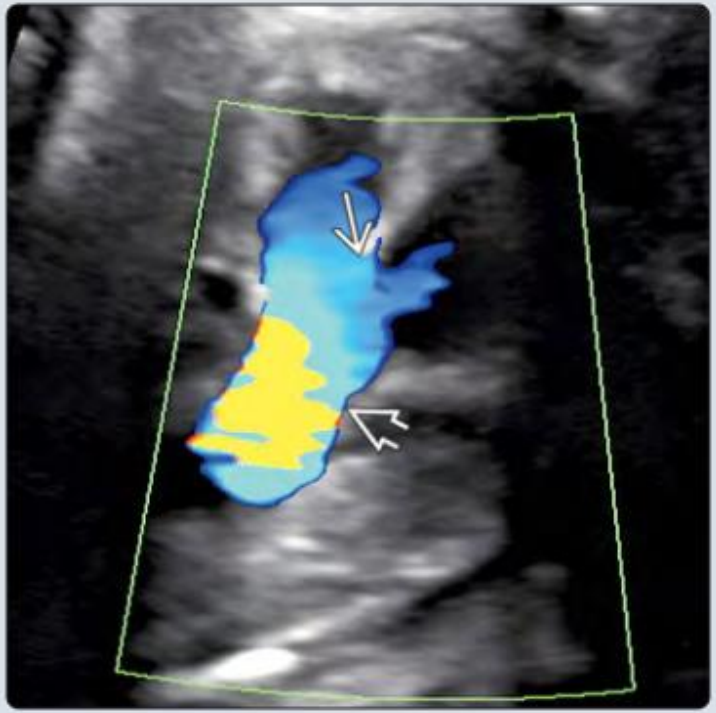


FIG 13-46 Diagrams and sonographic images of muscular ventricular septal defects (VSDs). The VSDs are indicated by the arrows. The first diagram (**A**) and the images in **C** and **D** show a midmuscular and apical muscular VSD. The second diagram (**B**) shows two midmuscular defects, and **E** shows an anterior muscular defect. Ao, aorta; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. (**A,** Adapted with permission from American Institute of Ultrasound in Medicine: AIUM practice guideline for the performance of fetal echocardiography. J Ultrasound Med 32:1067-1082, 2013.)

(Left) *Ultrasound shows a membranous VSD ⇨ with aortic override ⇨ in a fetus with tetralogy of Fallot.* **(Right)** *Color Doppler ultrasound in the same plane shows flow ➡ across the VSD from the RV and LV exiting the aorta ➡. Approximately 1/2 of all VSDs are associated with other cardiac malformations.*





VSD

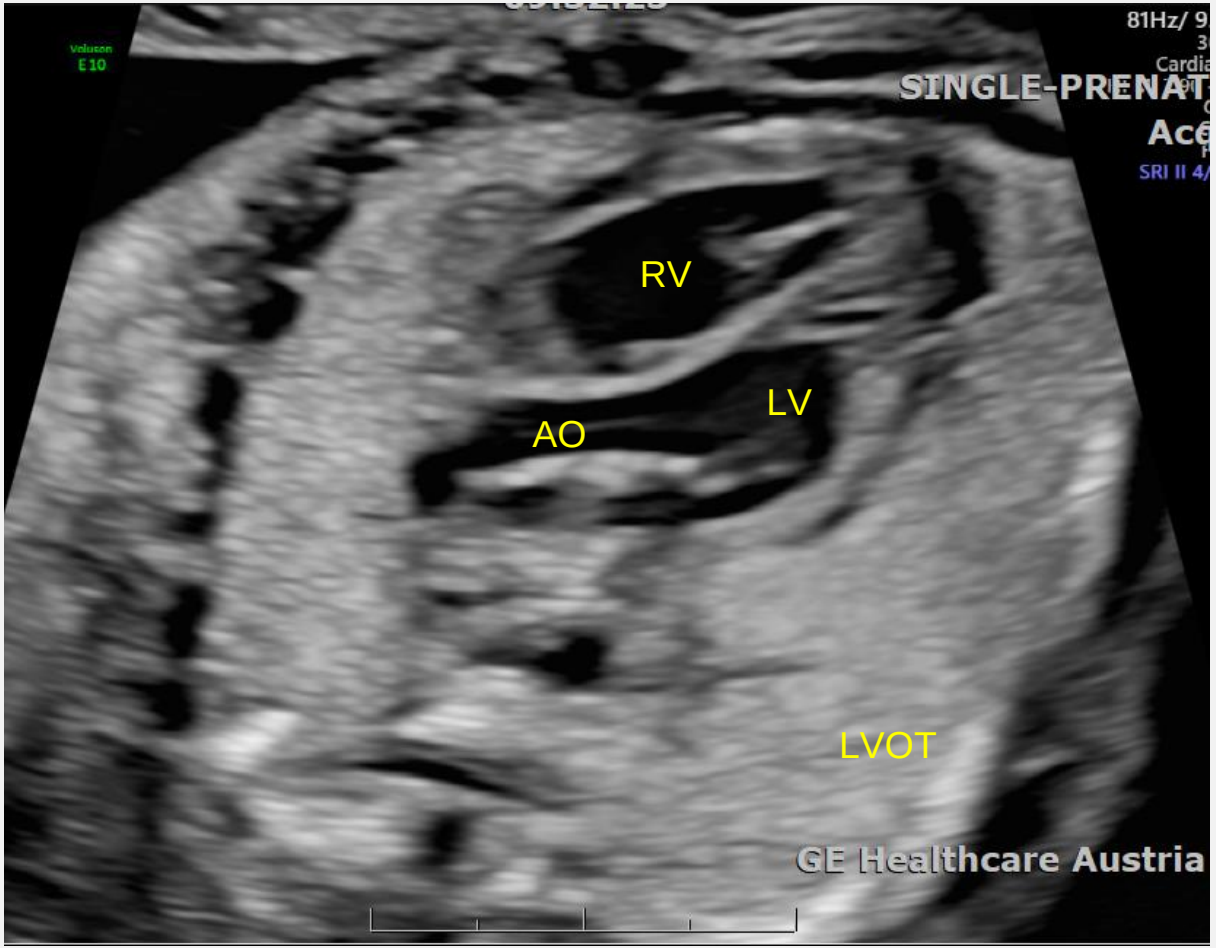
- **Recommendations**
- **Genetics**
  - Chromosomal anomaly found in > 40%
  - Offer karyotype if complex congenital heart disease or extracardiac abnormalities

Case review

01

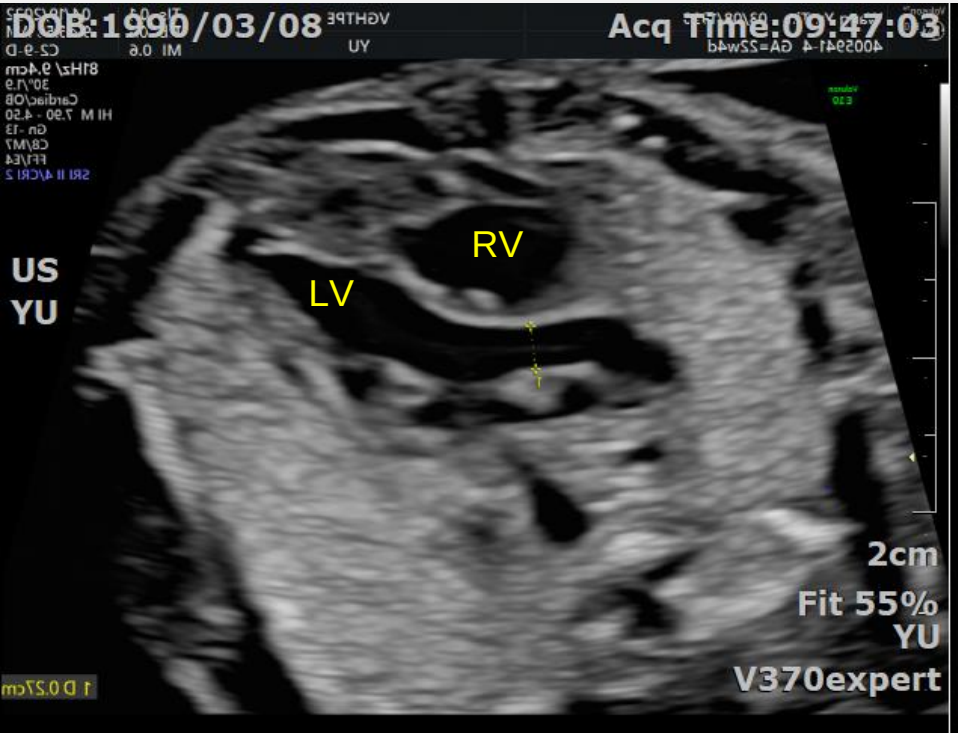
32 y/o

Pregnancy
22+4 weeks



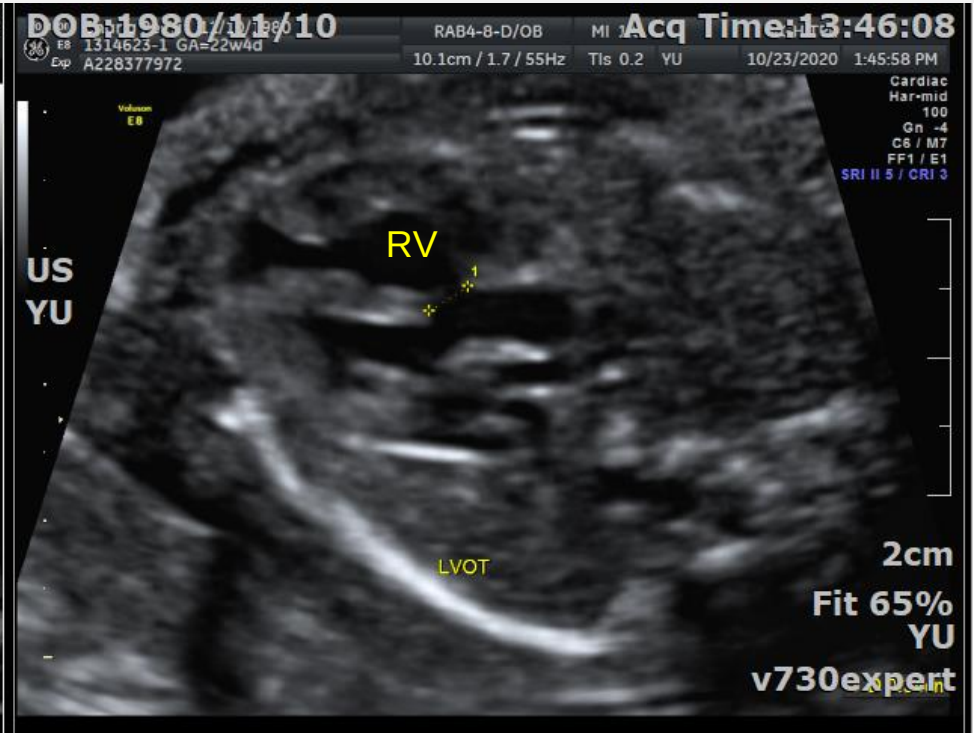
02

Pregnancy 22+4 weeks, 32 y/o



Pregnancy 22+4 weeks, 39y/o

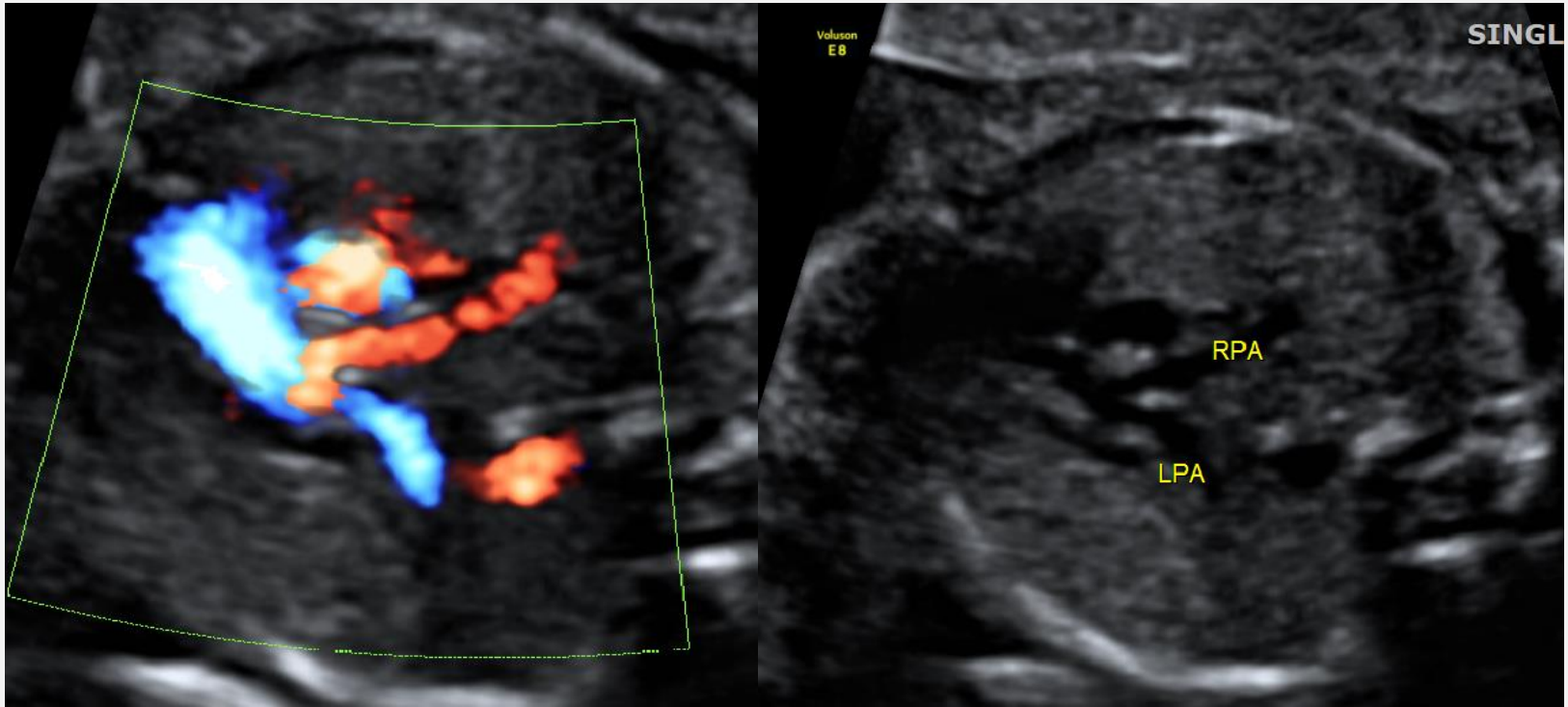
- Overriding aorta
- VSD 3.4mm



02

Pregnancy 22+4 weeks, 39y/o

- Overriding aorta
- VSD 3.4mm
- No obvious pulmonary stenosis noted



02

Pregnancy 22+4 weeks, 39y/o

- Overriding aorta
- VSD 3.4mm

- 20201112 aCGH array:

- Chip information: Affymetrix Cytoscan 750K array SA204136

- Result: arr (1-22)x2,(X,Y)x1
  No pathologic gene dosage variation (CNV) detected
  No pathologic absence of heterozygosity (AOH)

02

Pregnancy 22+4 weeks, 39y/o

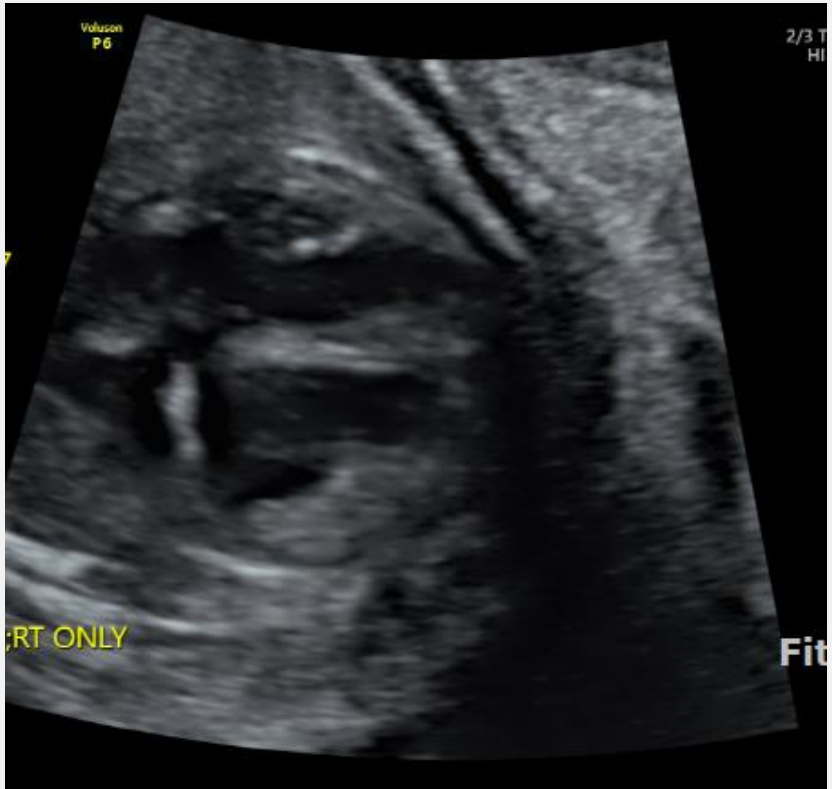
- Overriding aorta
- VSD 3.4mm

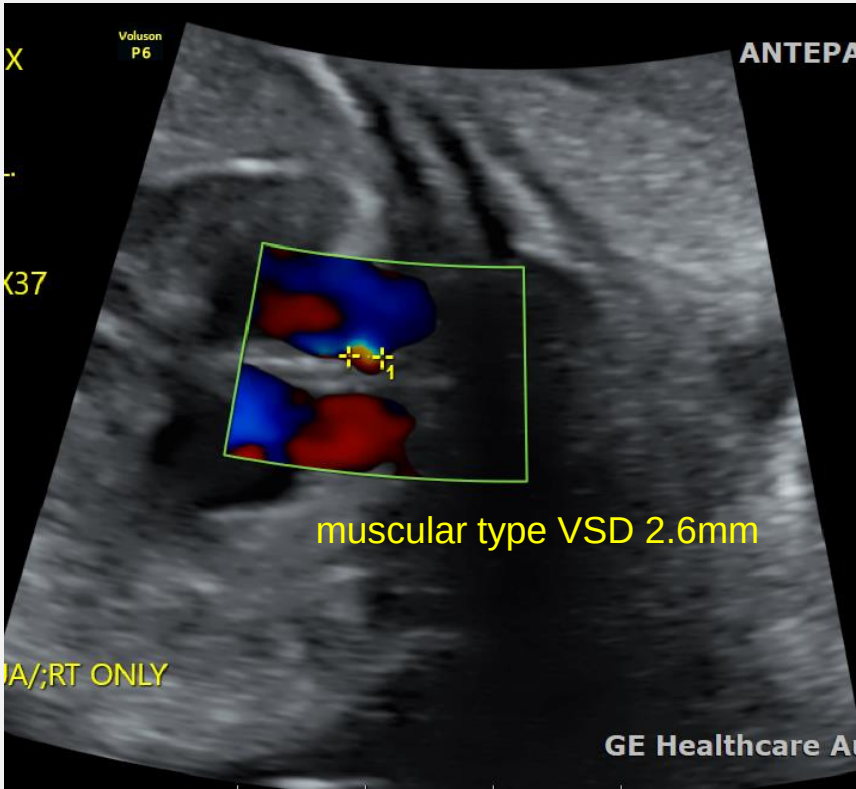
Pregnancy 36+1 weeks, preterm labor
- s/p vaginal delivery on 2021/01/26

- Type 2 ventricular septal defect,
  with regular Digoxin and Captopril, Synagis (Palivizumab)

03

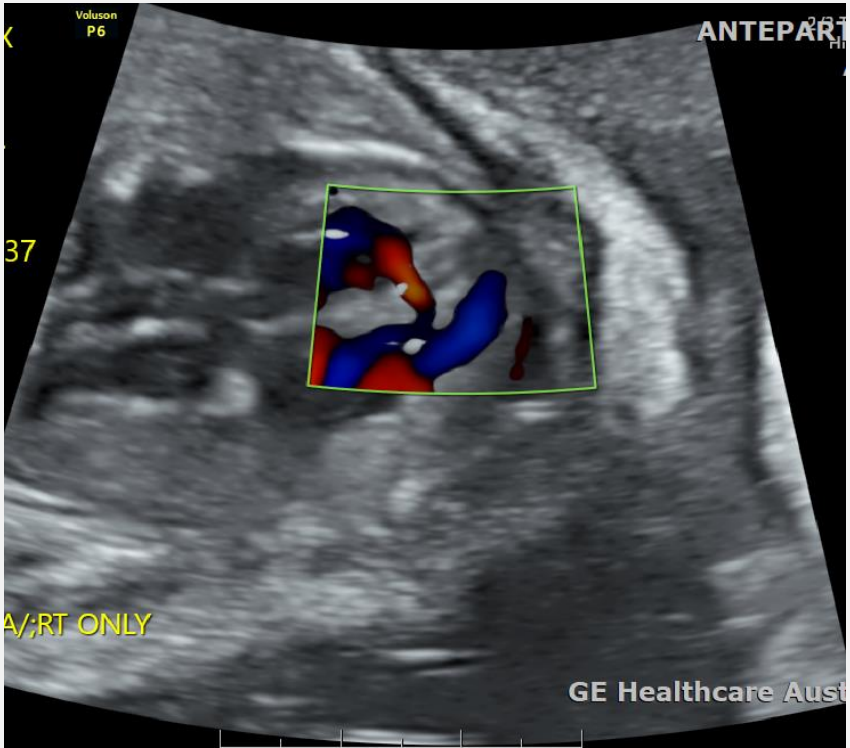
Pregnancy 30+0 weeks, 38 y/o

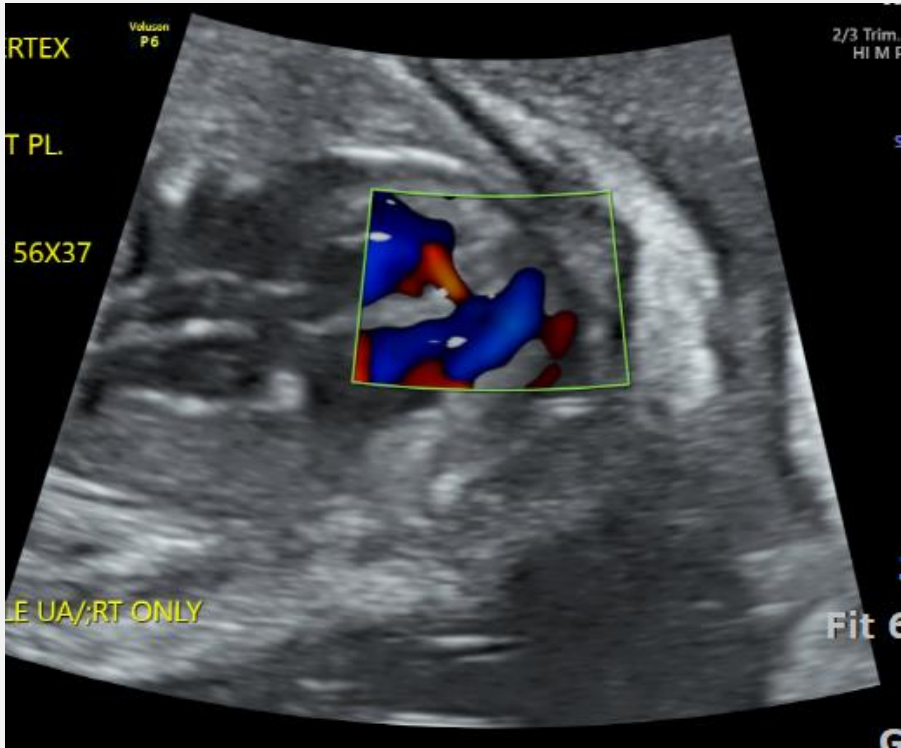




03

Pregnancy 30+0 weeks, 38 y/o





03

Pregnancy 30+0 weeks, 38 y/o

- Muscular type VSD 2.6mm

- **aCGH at Dr. Ko:**
- **arr 22q11.21q11.22(21,759-520-22,905,068)x1 (De novo)**, penetrance <10%
- Parental aCGH: both ok

03

Pregnancy 30+0 weeks, 38 y/o

- Muscular type VSD 2.6mm

Pregnancy 35+6 weeks, with PPROM and previous Cesarean section history
- s/p Cesarean section on 2020/06/14

THROAT & MOUTH
**High arch palate, bifid uvula

HEART ECHO:
Diagnosis : Case of **r/o 22q11 deletion anomaly**

Mild TR. No MR/AR/AS/PR/PS/CoA/TAPVR
Muscular VSD, two jets, around 0.15-0.2cm each. L-to-R shunt.
PFO, 0.26cm, L-to-R shunt. PDA closing.
Left side arch with normal branching pattern
Normal LV systolic function.

03

Pregnancy 30+0 weeks, 38 y/o

- Muscular type VSD 2.6mm

Pregnancy 35+6 weeks, with PPROM and previous Cesarean section history
- s/p Cesarean section on 2020/06/14

2021/09/28, 1Y3M

GR.II-III/VI SM OVER LSB
DOPPLER: M. VSD 0.195CM
NO CHF SIGN

04

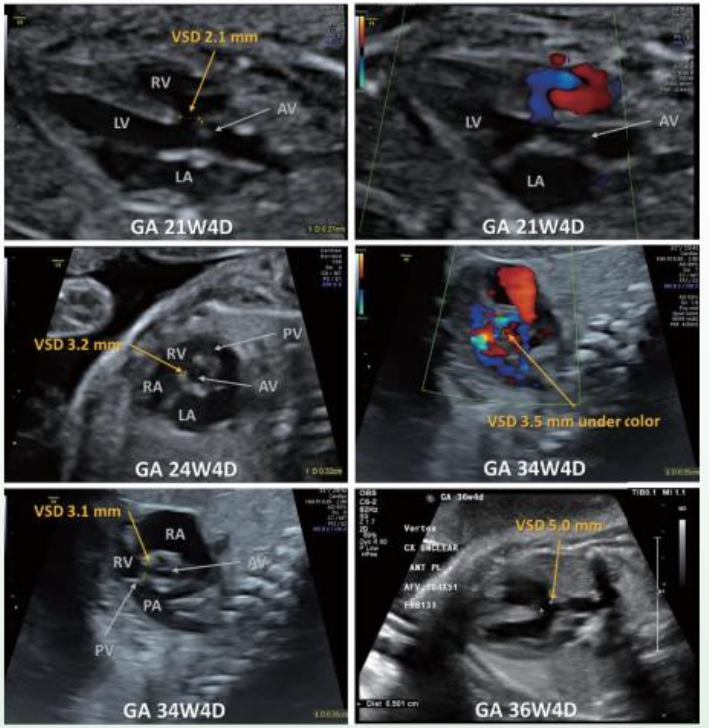
產前診斷單純心室中膈缺損之病例報告與文獻回顧

臺北榮民總醫院 婦女醫學部 陳冠宇醫師/葉長青醫師

病例報告

張女士，37歲，G1P0，過去無其他特殊病史。本次為自然懷孕，初期產前檢查均正常，第二孕期接受羊膜穿刺檢查顯示染色體套數正常，孕婦在妊娠21週接受高層次超音波檢查，發現胎兒心室中膈缺損(Ventricular septal defect, VSD)，其他無顯著結構異常。後續產檢追蹤情況良好，並無額外併發症出現。此孕婦於39週順利生產，胎兒體重3052克，身長48.4公分，出生後第一分鐘與第五分鐘的Apgar Score分別為8與9分。新生兒出生以後接受心臟超音波追蹤，診斷為Type II 心室中膈缺損7.1mm，合併雙向血流分流(Bidirectional shunt)與開放動脈導管(PDA)1.7mm。新生兒出院前再接受一次心臟超音波檢查，顯示無鬱血性心臟情況，改門診追蹤治療。新生兒後續每個月接受心臟超音波追蹤，顯示VSD有慢慢縮小的趨勢，最後一次追蹤為出生後1歲10個月，超音波顯示心室中膈缺損面積縮小為4.2mm，且無鬱血性心衰竭情況。 < 圖一 >

圖一：產前超音波診斷



References

- Norton, M. E. (2016). Callen's Ultrasonography in Obstetrics and Gynecology.
- Woodward, P. J., Kennedy, A., & Sohaey, R. (2016). Diagnostic Imaging: Obstetrics (3rd ed.). Philadelphia, PA: Elsevier - Health Sciences Division
- Wyman W. Lai, Luc L. Mertens, Meryl S. Cohen, Tal Geva. Echocardiography in Pediatric and CHD - 2nd edition 2016
- 臺灣周產期醫學會會訊 – 2021/10
- Bravo-Valenzuela, N. J., Peixoto, A. B., & Araujo Júnior, E. (2018). Prenatal diagnosis of congenital heart disease: A review of current knowledge. *Indian heart journal*, *70*(1), 150–164.
- Lee, W., & Comstock, C.H. (2006). Prenatal Diagnosis of Congenital Heart Disease: Where Are We Now? *Ultrasound Clinics, 1*, 273-291.

Thank you !