Atrioventricular Septal Defect Fetal Imaging: a spectrum of disease

Professor Gurleen Sharland

Atrioventricular Septal Defect

Common congenital heart lesion ~ 5%

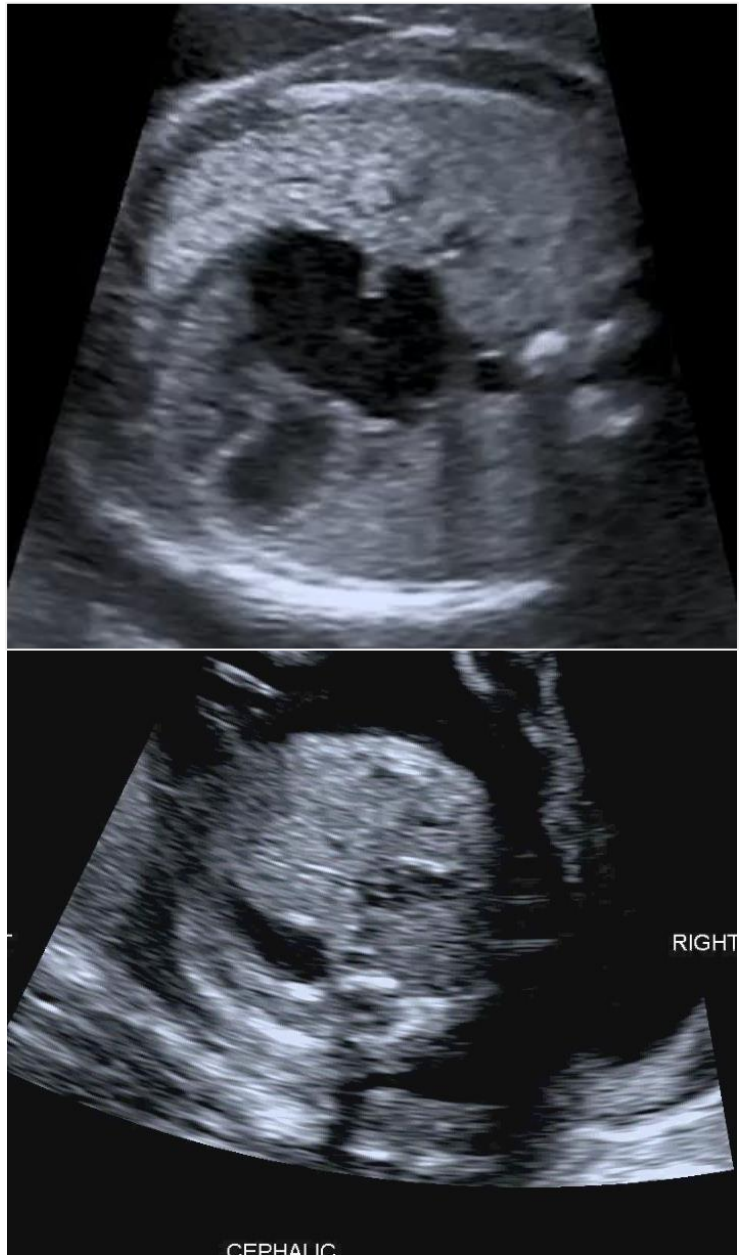
Common finding in fetal series ~ 10%

Detectable by four chamber view screening

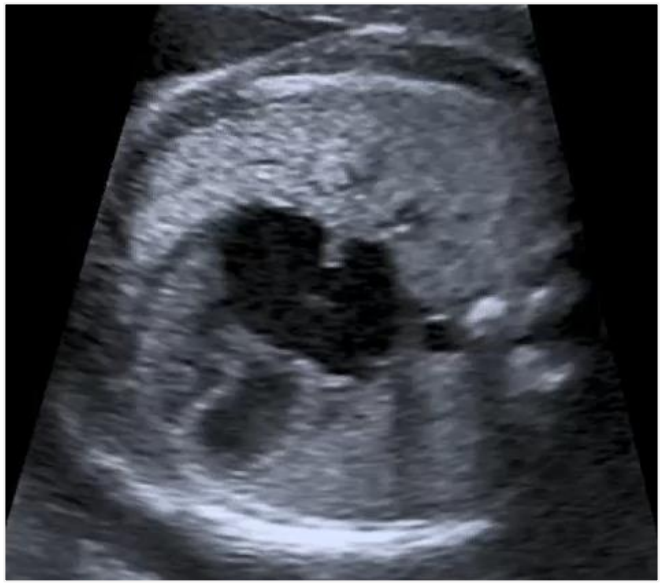
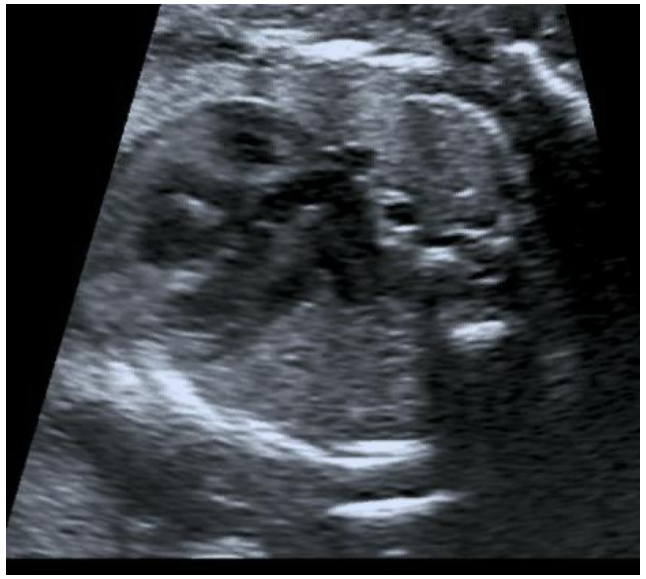
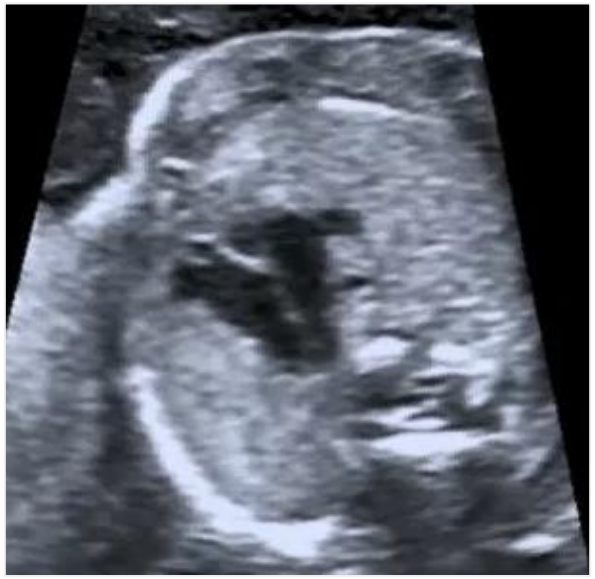
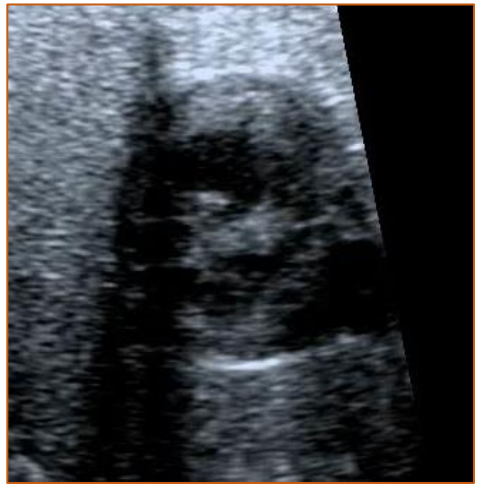
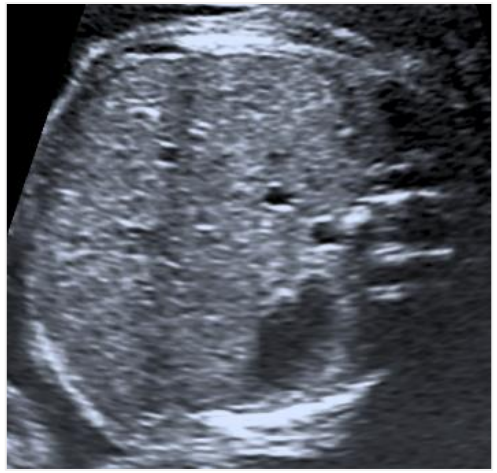
- Loss of differential insertion of AV valves
- No offsetting of valves (Sonographers taught to examine crux of heart and look for offsetting)
- Defect in atrial/ventricular septum identified

Referral reasons from screening:

- Abnormal four chamber view
- Increased nuchal translucency
- High risk for Trisomy 21 on screening
- Extra-cardiac abnormality
- Malposition suggesting laterality defect



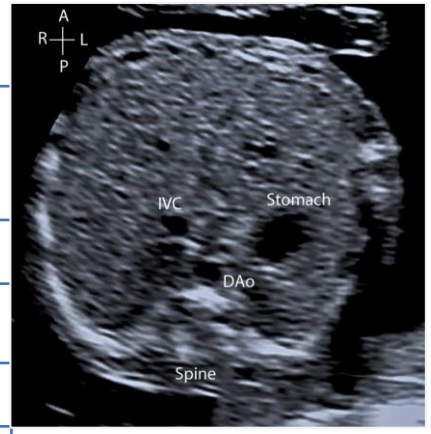
AVSD - Isolated
Normal situs, balanced ventricles, normally related great arteries

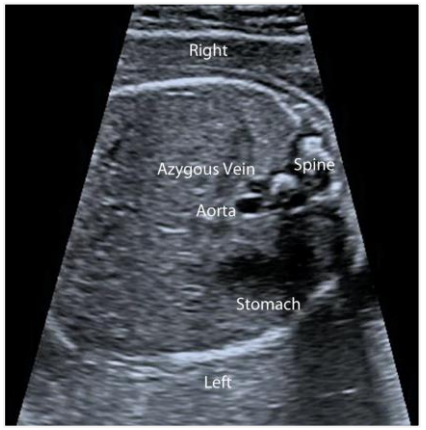
Atrioventricular Septal Defect

Normal situs

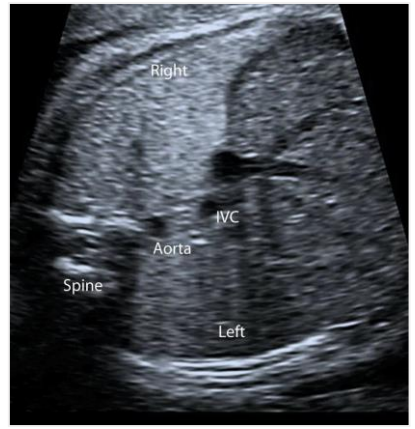
- Increased risk chromosomal abnormalities



Left atrial isomerism

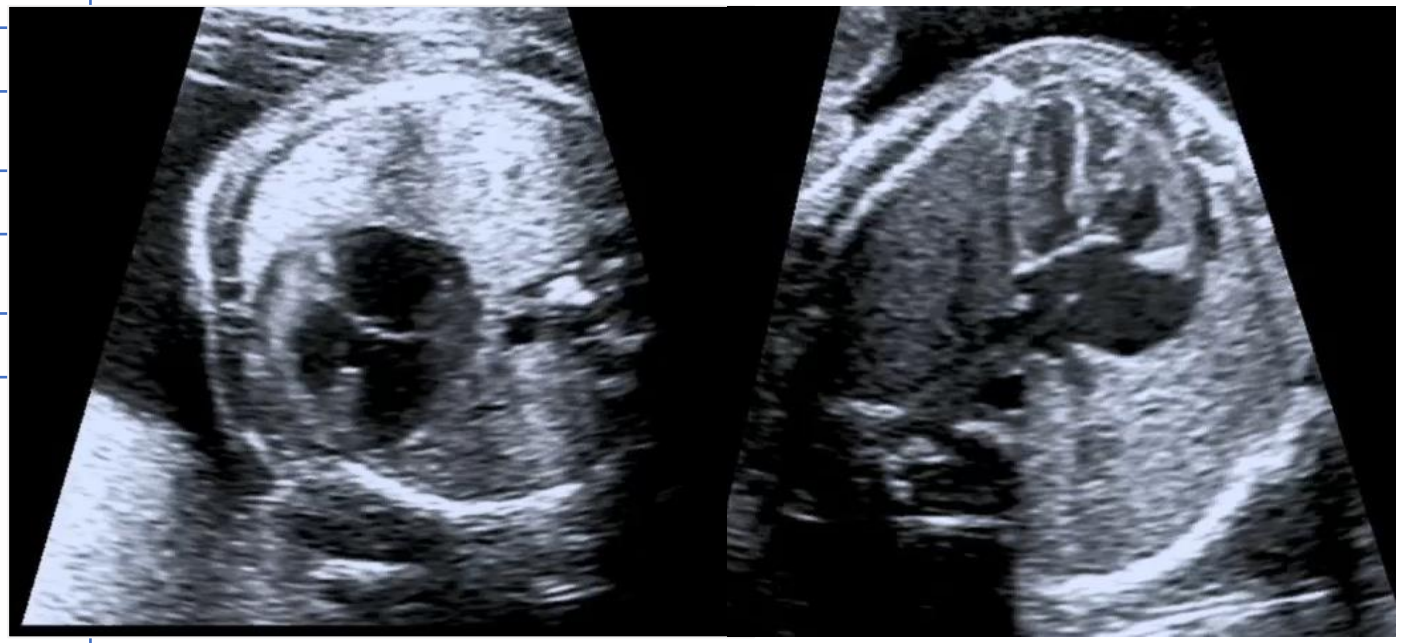


Right atrial isomerism



Ventricular balance/imbalance

Variable size atrial and ventricular components

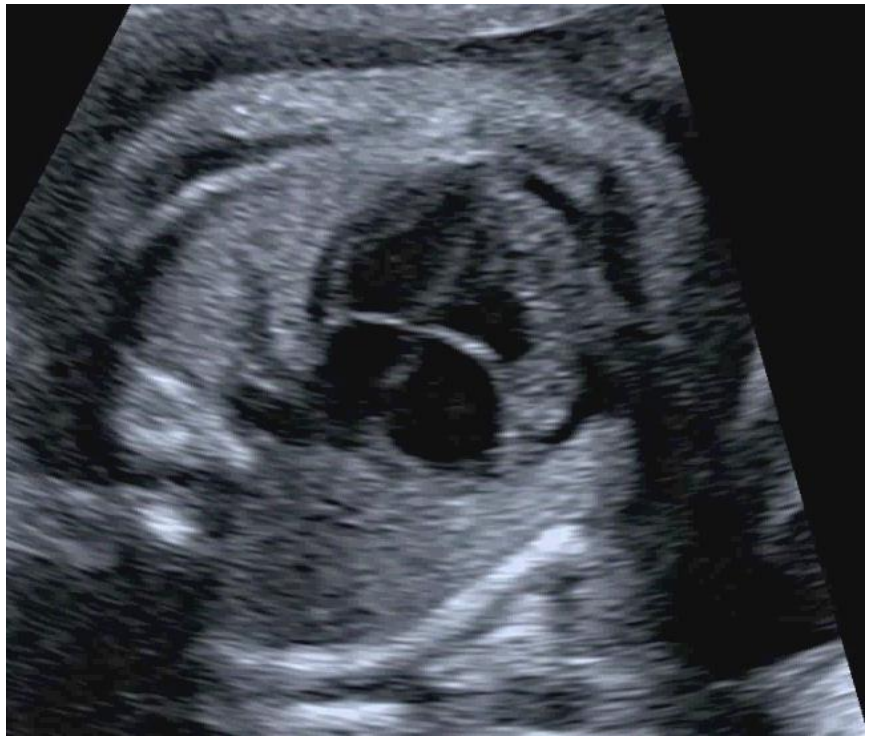


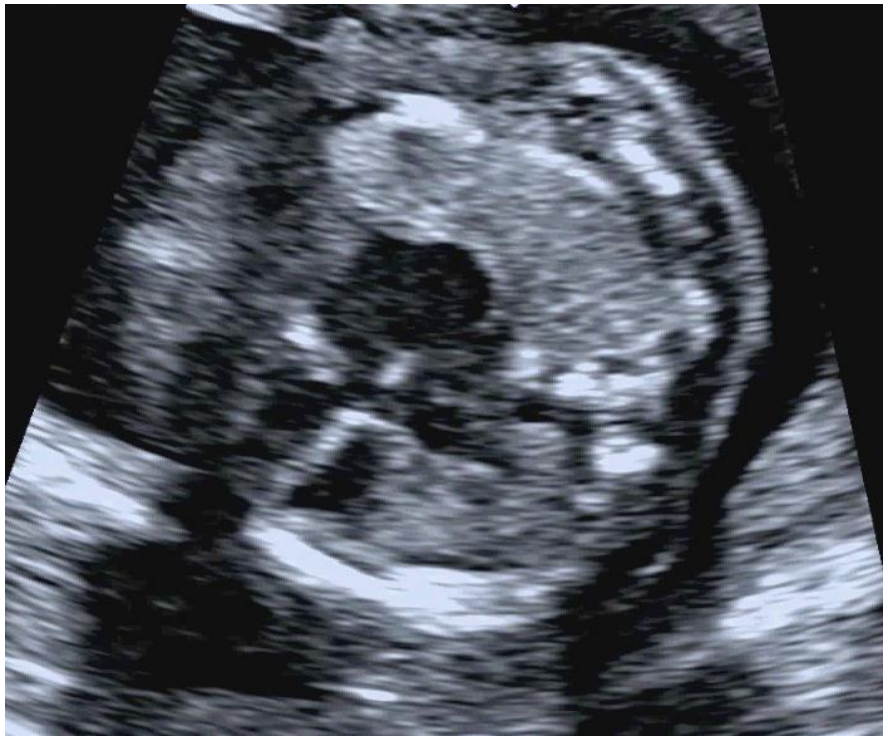
Additional cardiac abnormalities

- Coarctation of the aorta
- Tetralogy of Fallot
- Pulmonary stenosis/ atresia
- Bilateral SVC

Partial AVSD

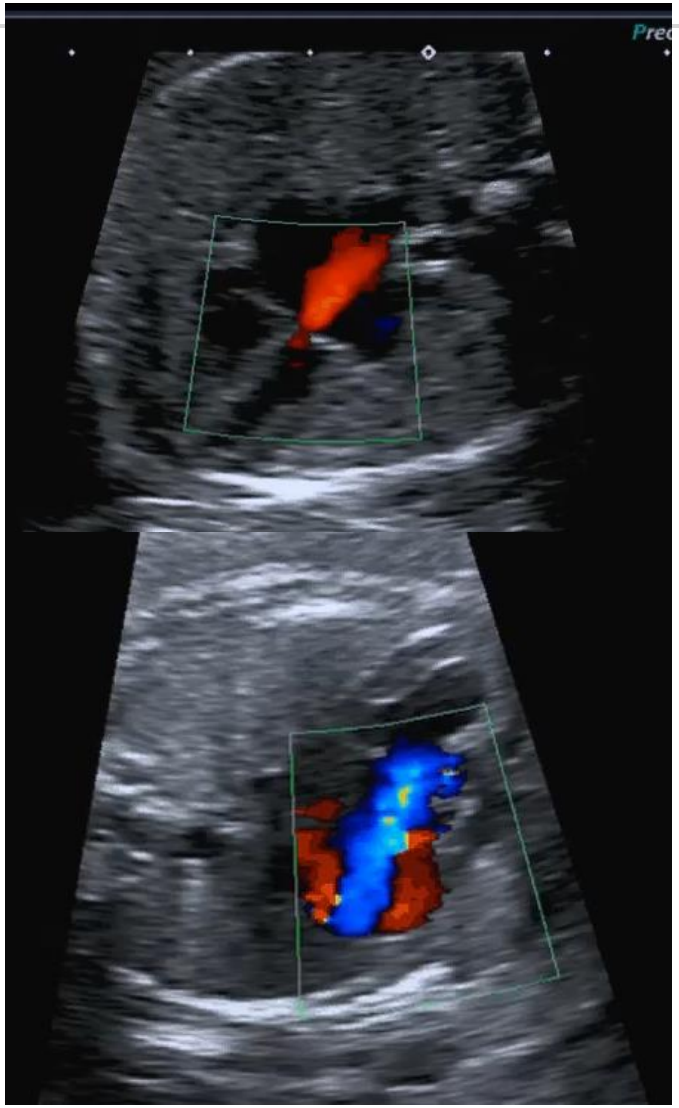
Can be more difficult to detect during obstetric screening





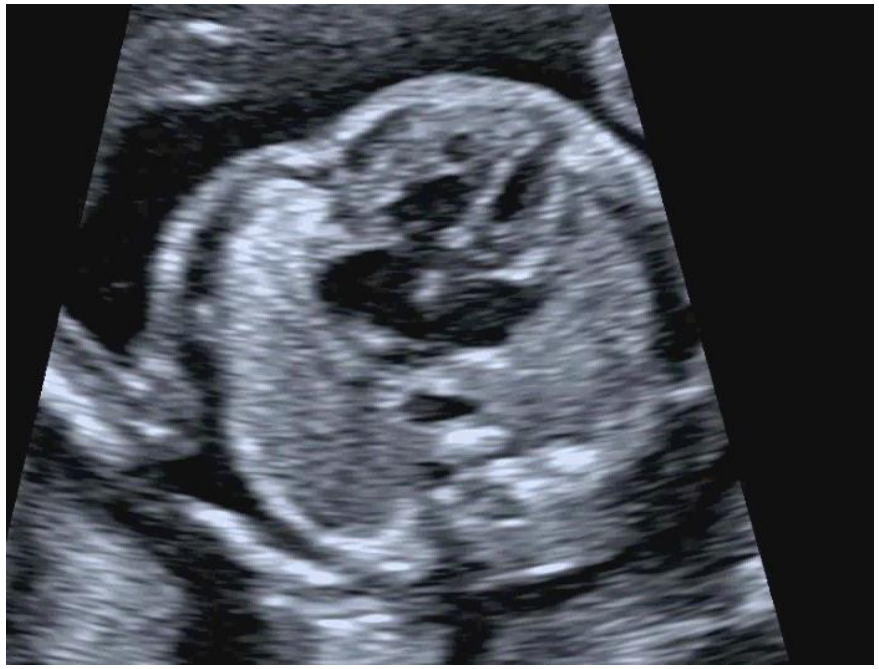
Adverse Prognostic Factors

- Atrioventricular valve regurgitation
  - Can lead to fetal hydrops if severe
- Ventricular imbalance
- Other associated CHD
  - Coarctation, TAPVD, tetralogy
- Isomerism

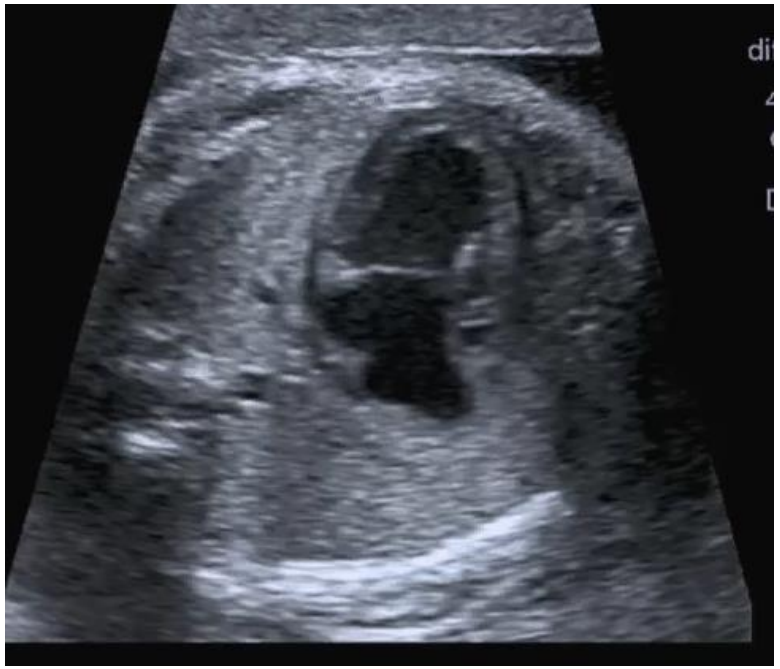


Ventricular Dominance

- Can be left or right dominant ventricle
- Varying degree of sizes LV or RV



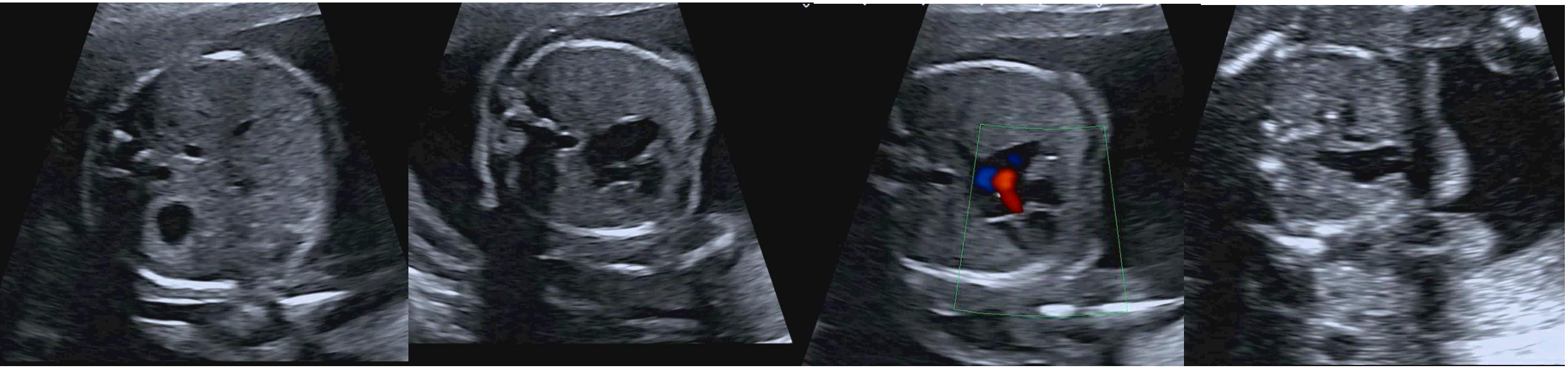
Mild LV Hypoplasia



Severe RV Hypoplasia

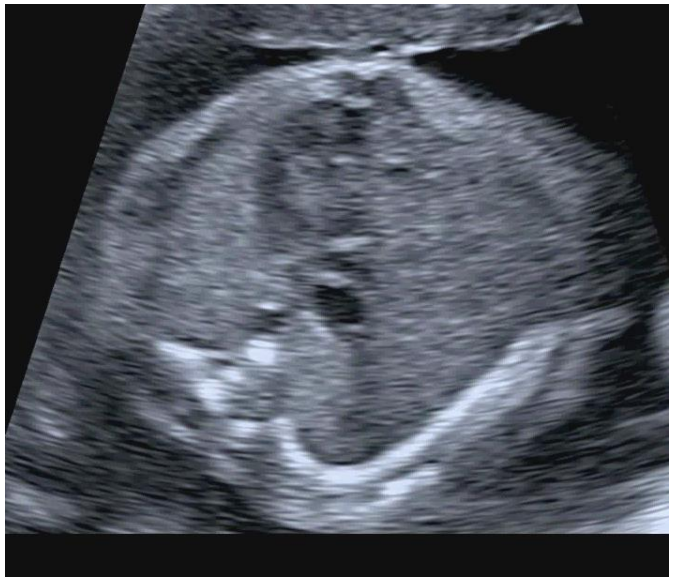
AVSD
Ventricular Dominance (RV)

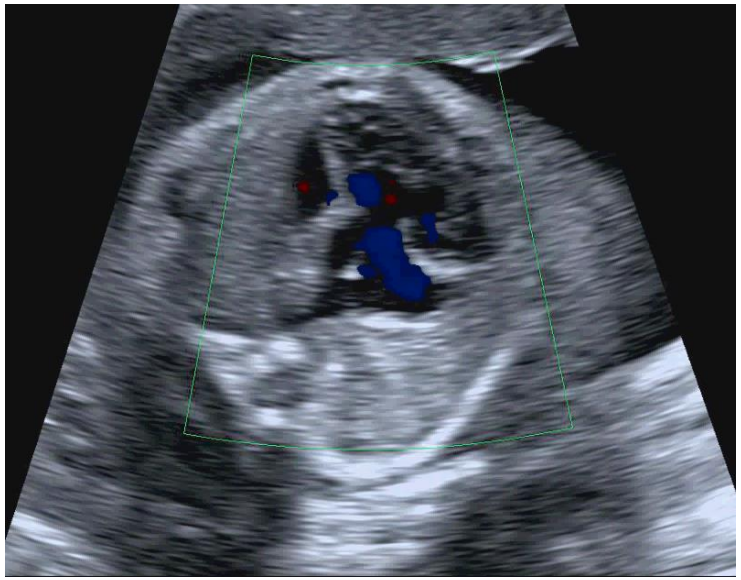
- Dominant RV
- Severe LV hypoplasia
- Hypoplastic aortic arch
- Falling into spectrum of HLHS

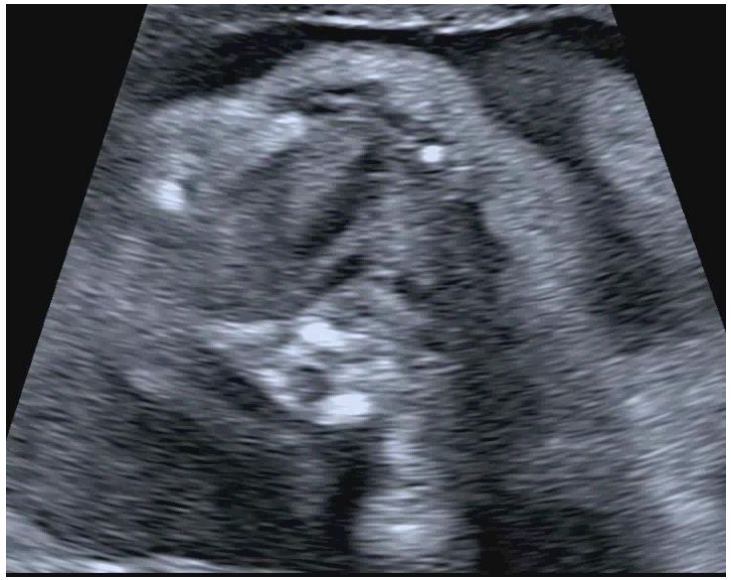


AVSD with coarctation

- Dominant RV, LV hypoplasia
- Slender aortic arch
- Small LV may recover after arch repair
- Biventricular circulation possible

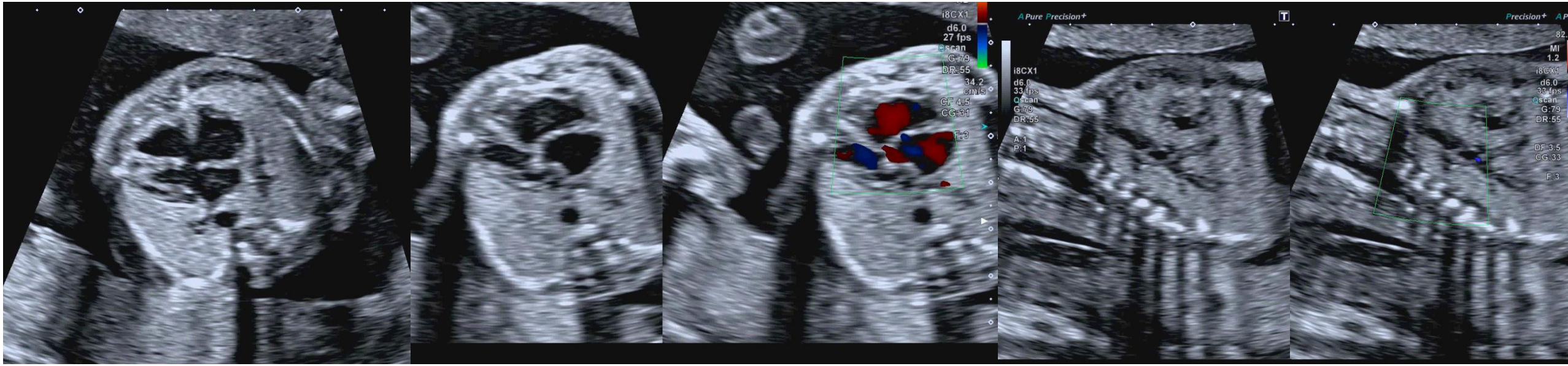






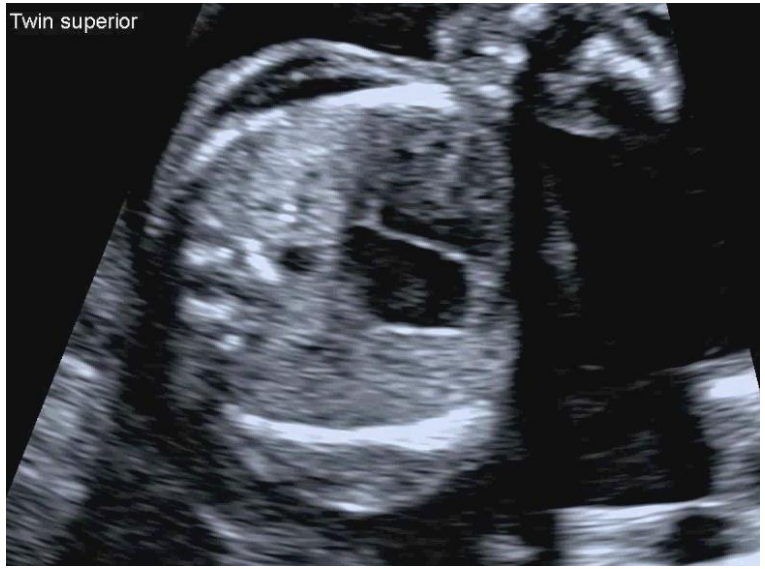
AVSD with coarctation

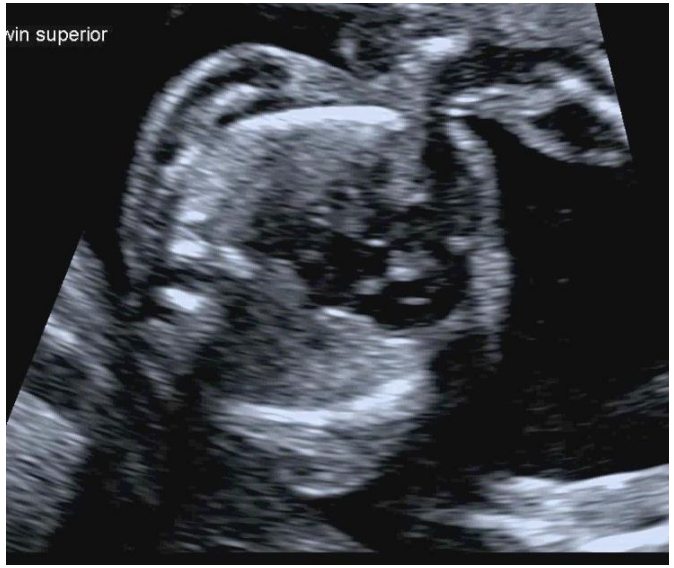
- Dominant RV, LV hypoplasia
- More complex
- AV valve regurgitation
- Management more challenging

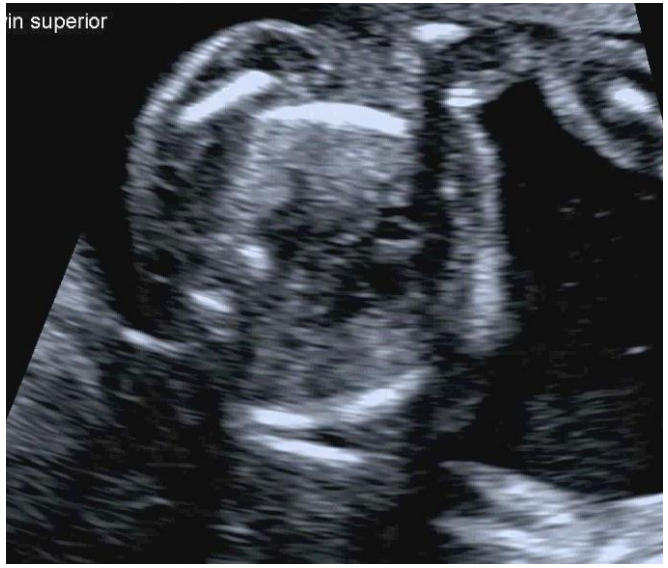


AVSD with Tetralogy of Fallot

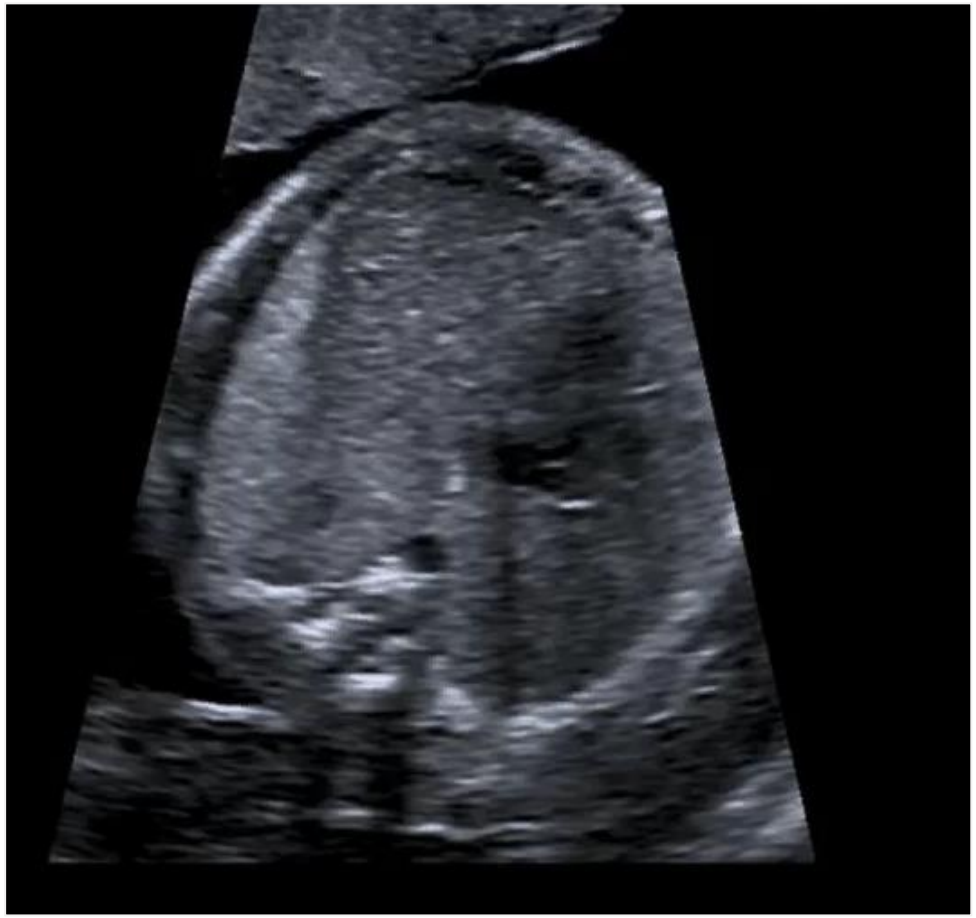
- Balanced ventricles
- Overriding aorta
- Small pulmonary artery
- High risk for trisomy 21

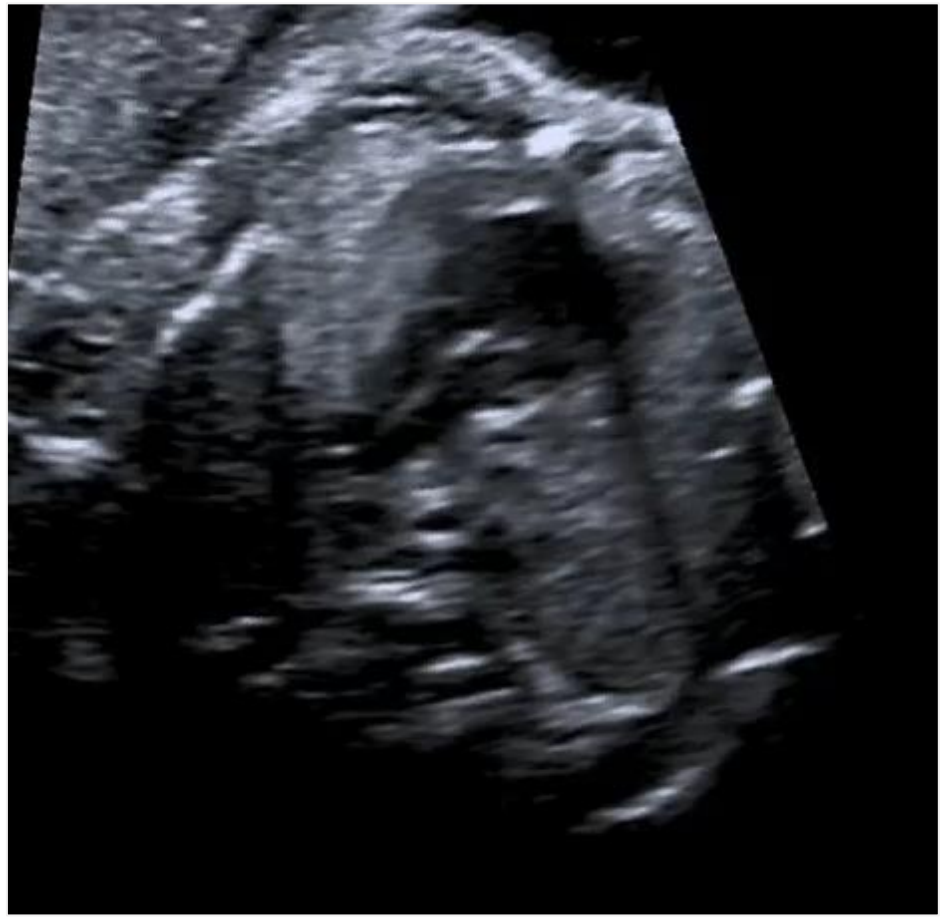






AVSD, Double Outlet Right Ventricle, Aorta anterior





AVSD with Left Atrial Isomerism (LAI)

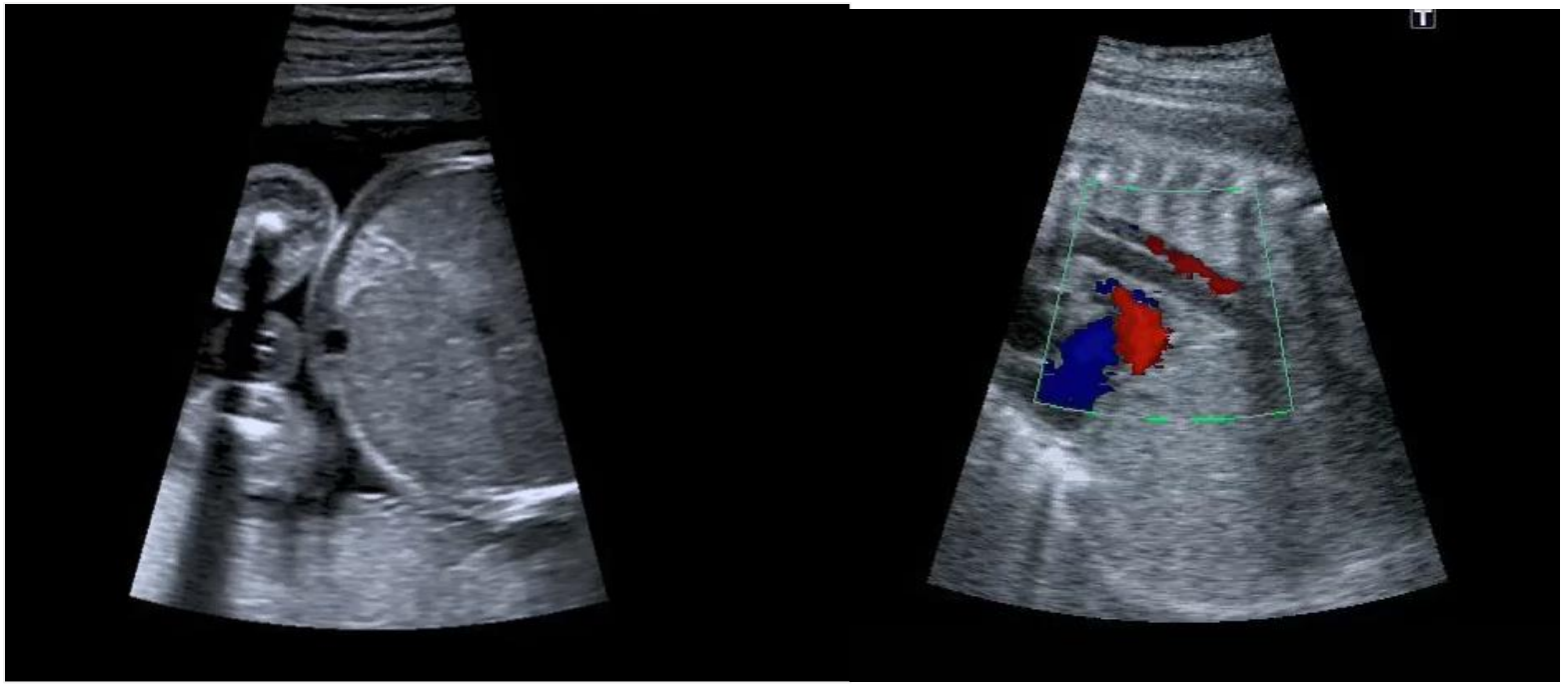
Features of LAI

- Abnormal cardiac position
- Heart and stomach on opposite sides
- Interrupted IVC
- Azygous continuation
- Bradycardia
- Cardiac defects
  - AVSD, coarctation, anomalous pulmonary venous drainage, bilateral SVC

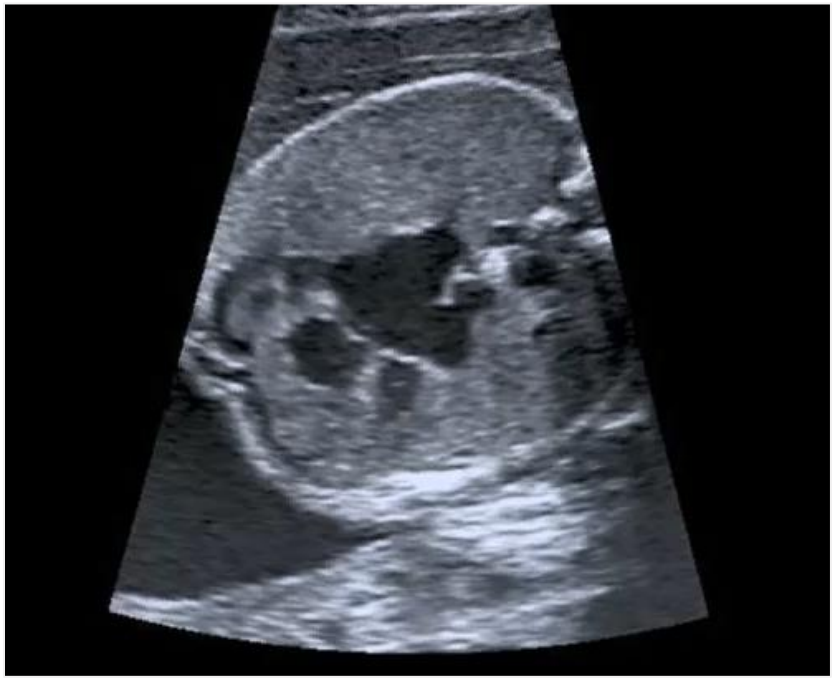
Things to also consider in LAI

- Polyspenia
- Congenital heart block
- Bowel malrotation
- Biliary atresia
- Chromosomal abnormalities rare
- Wide spectrum of abnormality

AVSD
with Left Atrial Isomerism (LAI)

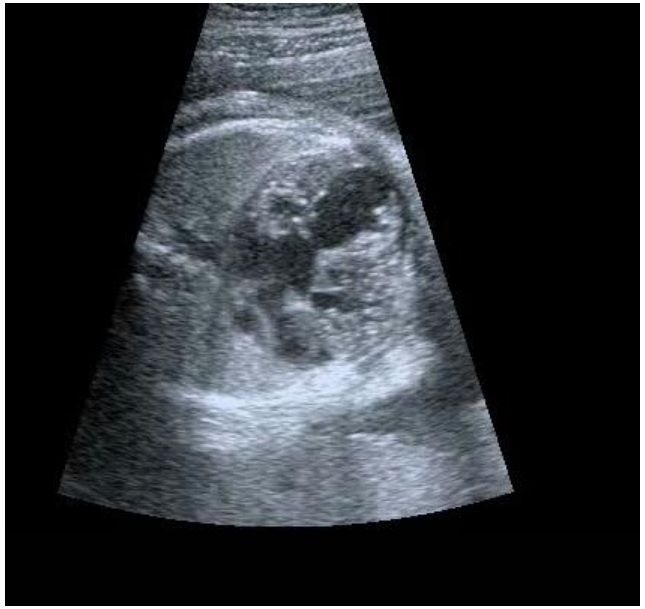


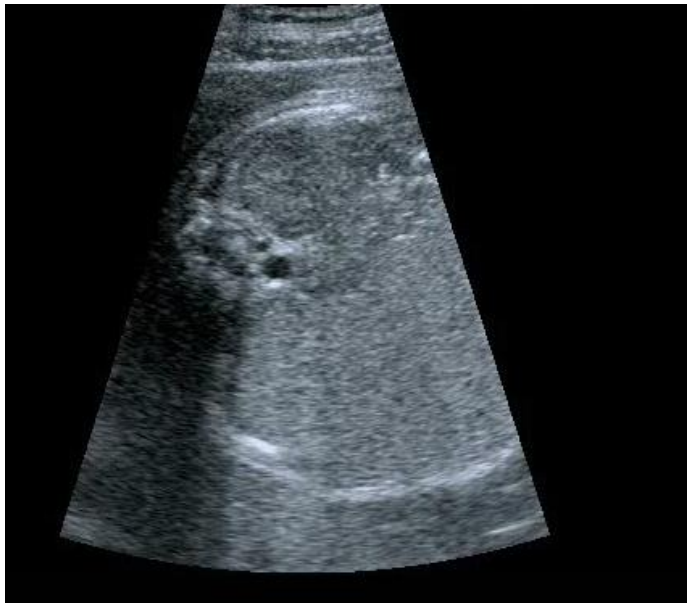
Interrupted IVC
Azygous continuation
LAI

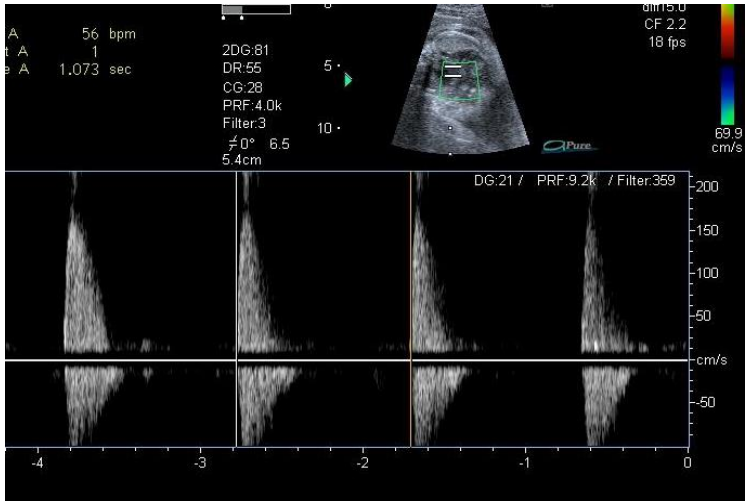


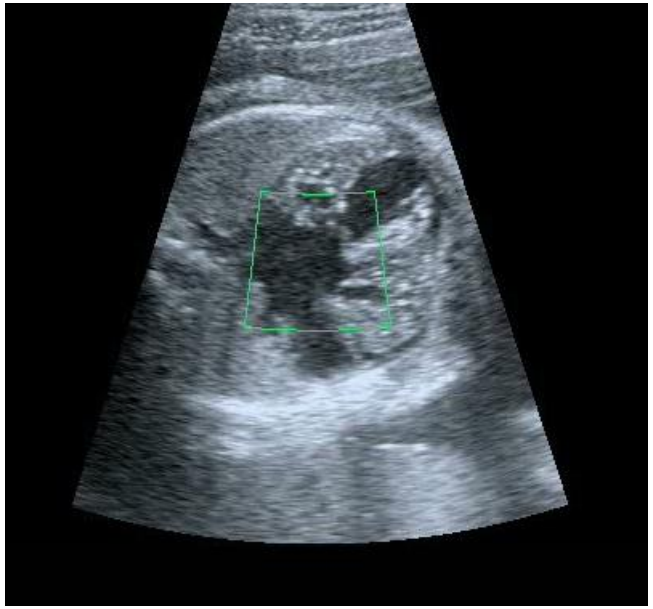
AVSD

AVSD, LAI,
Complete Heart Block









AVSD with Right Atrial Isomerism (RAI)

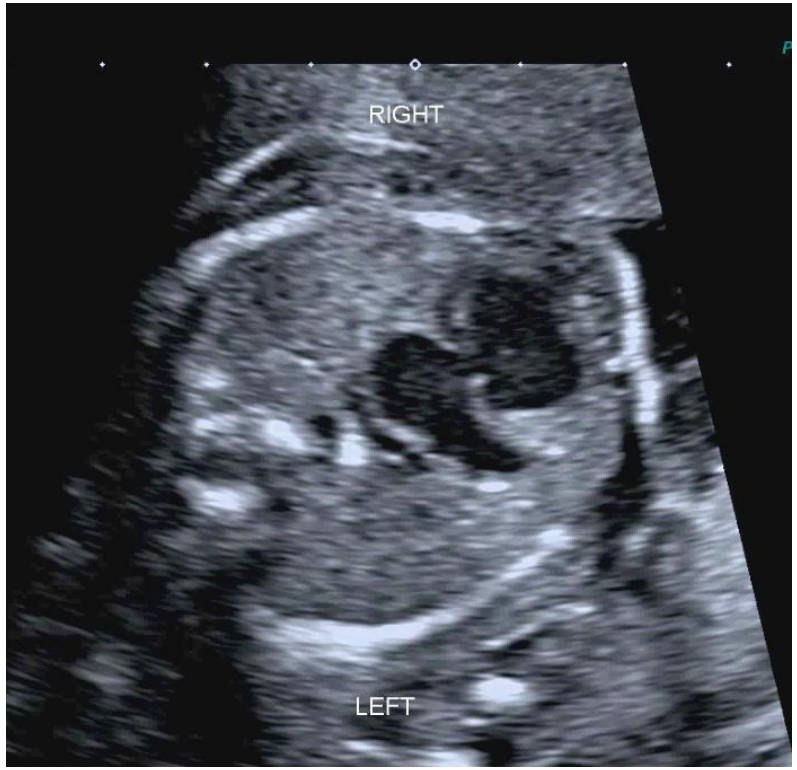
Features of RAI

- Abnormal cardiac position
- Heart and stomach on opposite sides
- IVC and DAo lie on same side of spine
- IVC lies directly anterior to aorta in abdomen
- Complex cardiac malformations
  - AVSD, DORV, pulmonary atresia/stenosis
- Anomalous pulmonary venous drainage

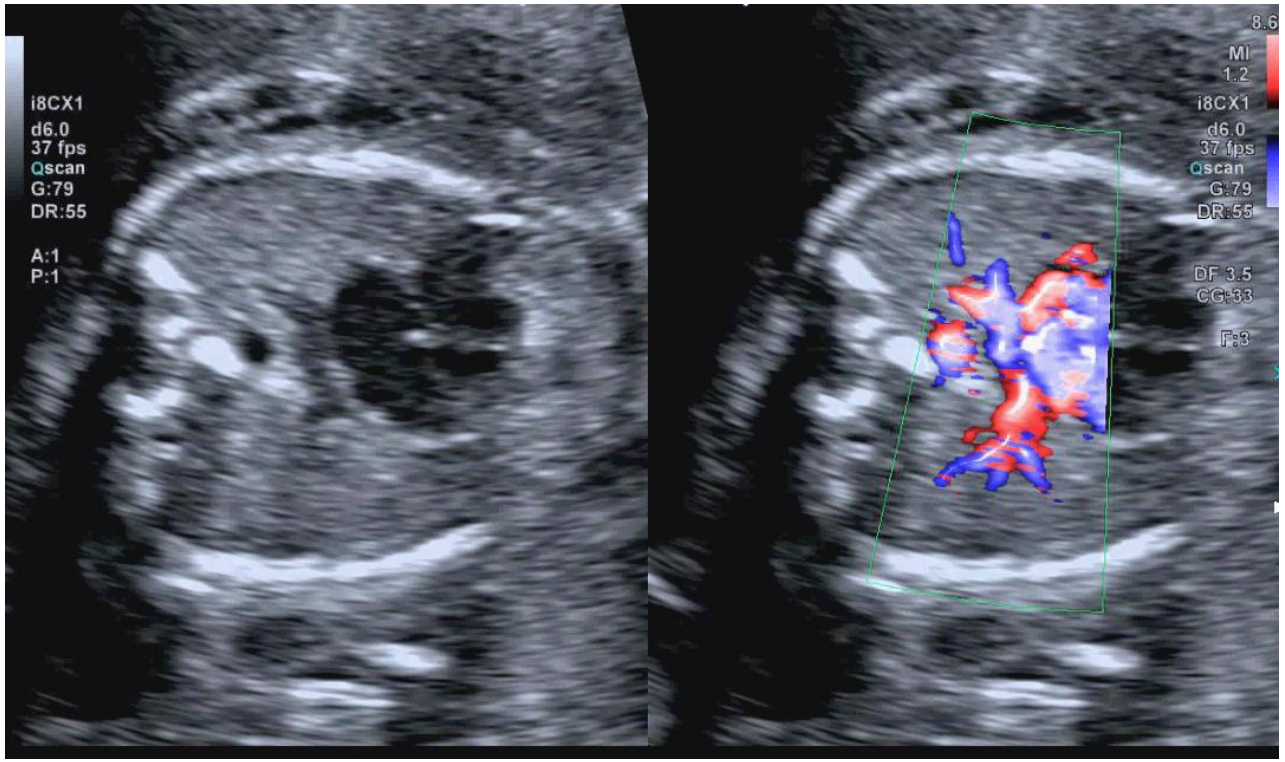
Things to also consider in RAI

- Asplenia
- Bowel malrotation
- Chromosomal abnormalities rare
- Wide spectrum of abnormality

Dextrocardia, RAI, AVSD,
Total Anomalous Pulmonary Venous Drainage (TAPVD)

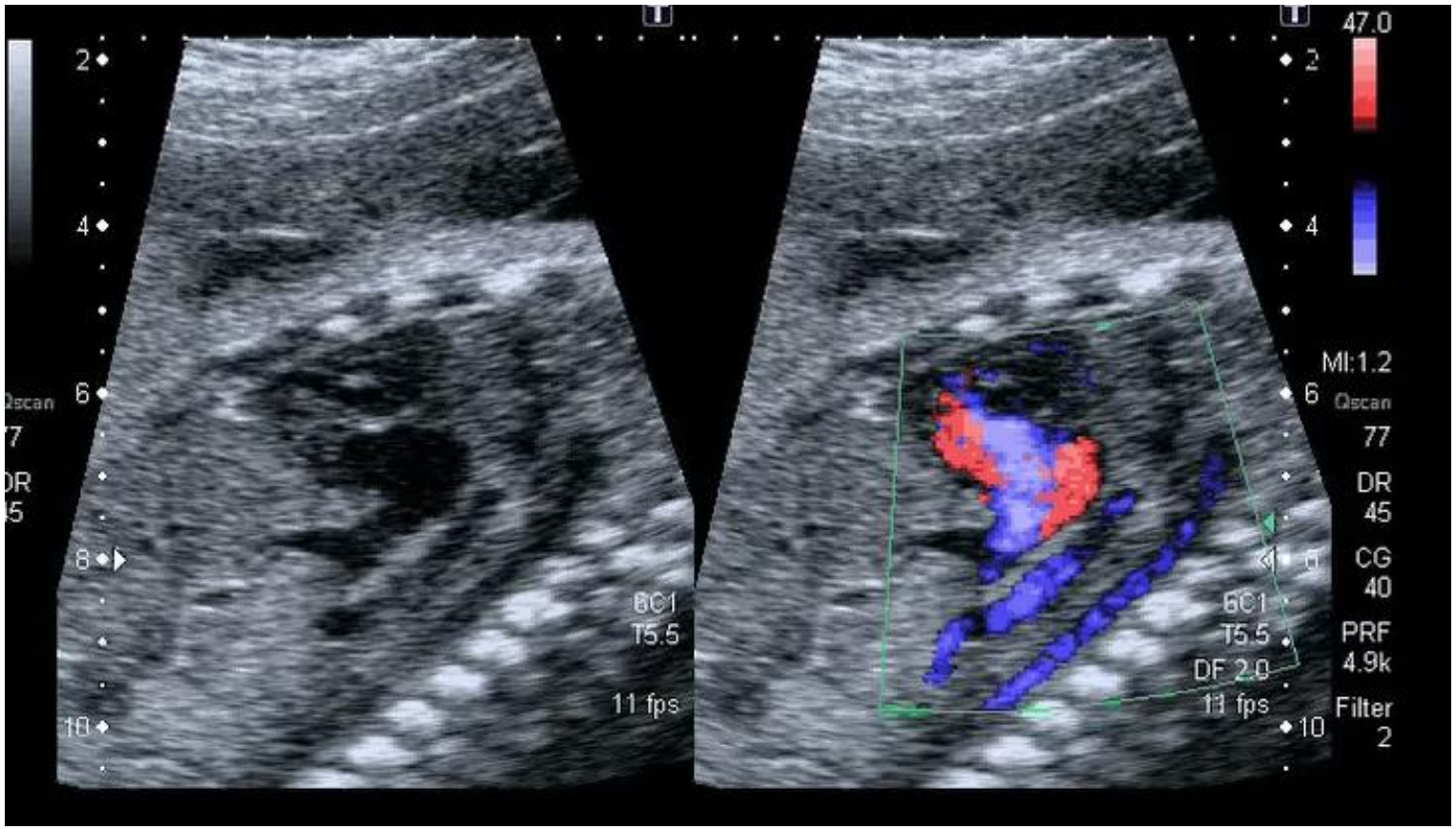


Unbalanced AVSD

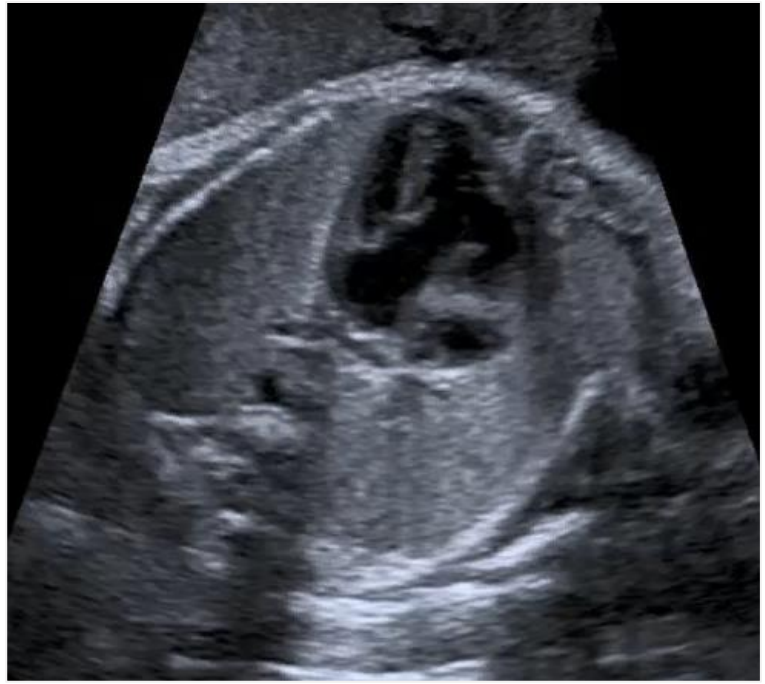


TAPVD

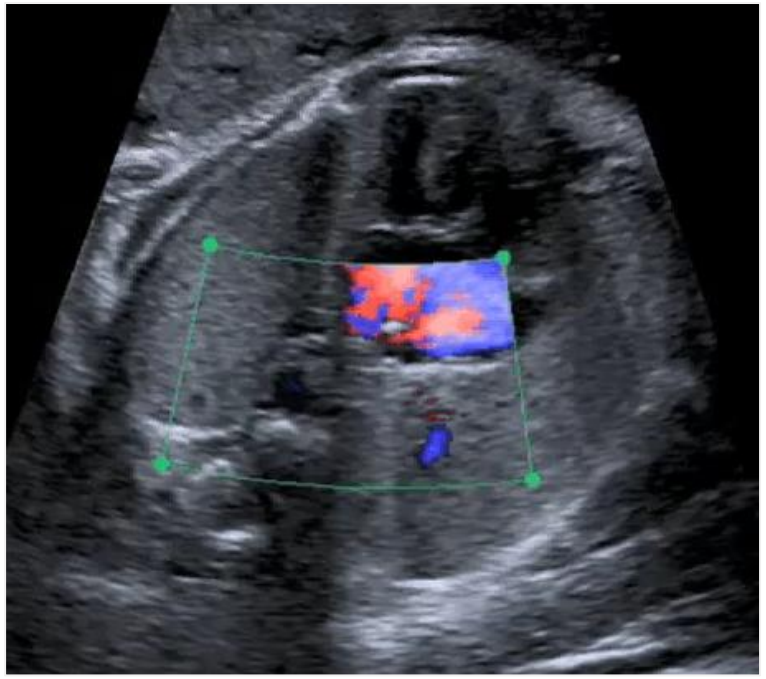
Infracardiac TAPVD in RAI



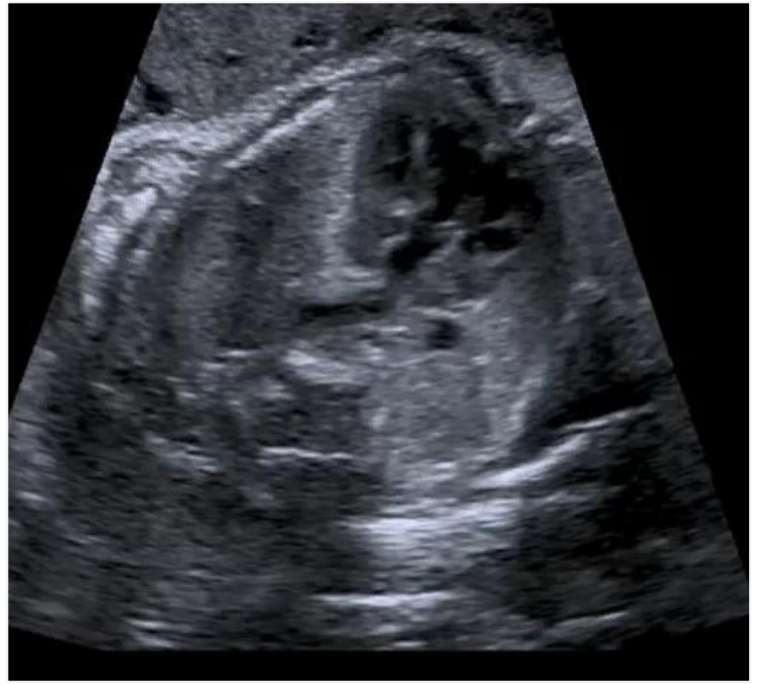
RAI , AVSD, DORV,
Total Anomalous Pulmonary Venous Drainage (TAPVD)



Unbalanced AVSD



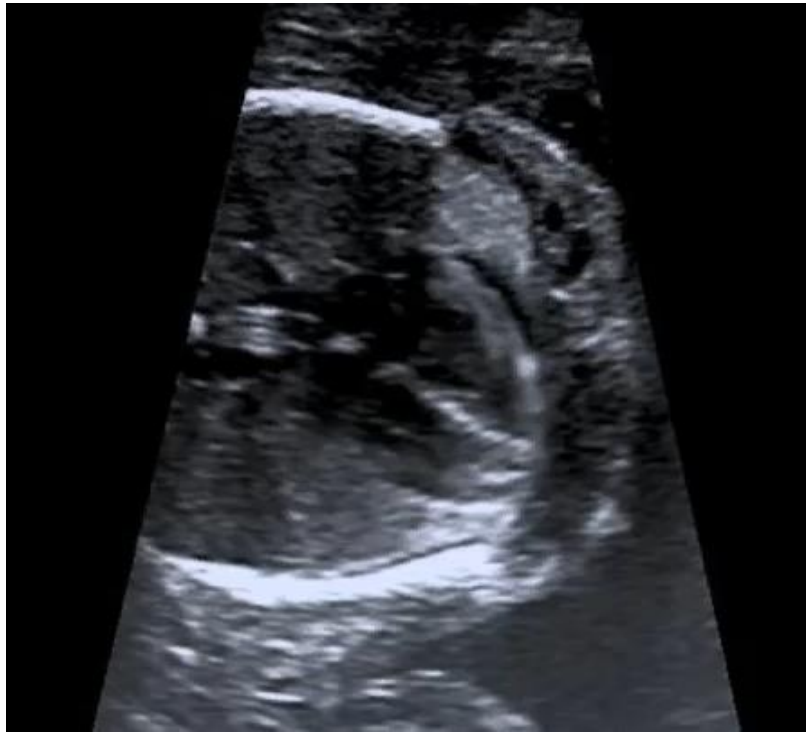
TAPVD

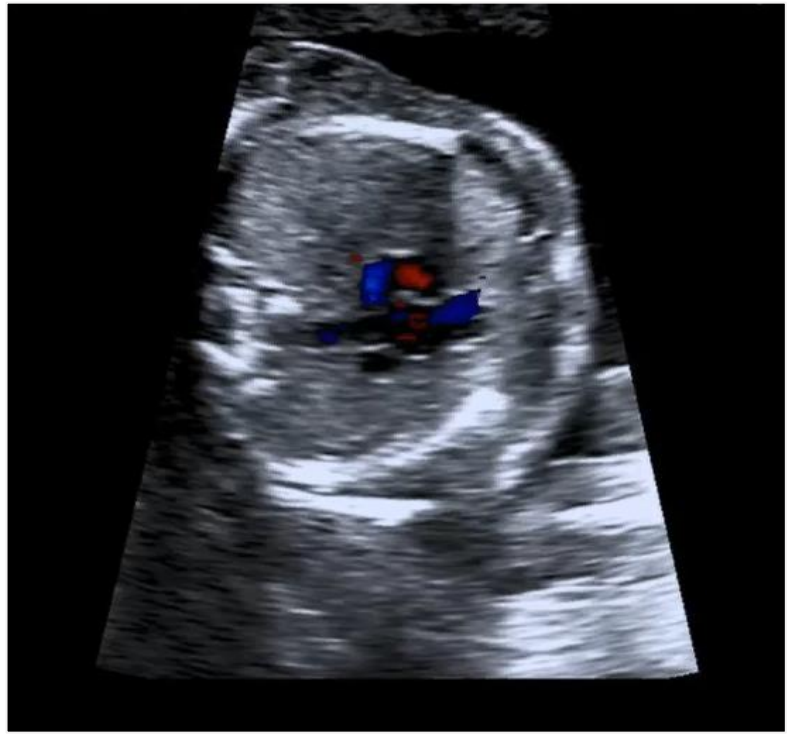


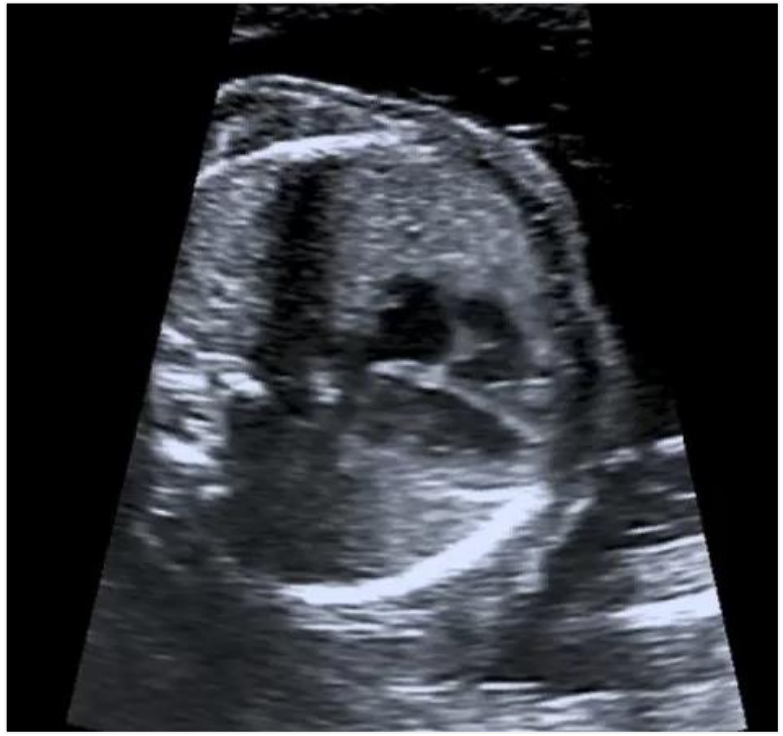
DORV

AVSD
Pitfalls in diagnosis

- Inlet VSD may appear to have lost AV valve offset
- LSVC to CS may be mistaken for AVSD
- Foreshortened 4 chamber view may be difficult to assess







Counselling and Management

Spectrum of abnormality

Fetal echocardiography allows accurate diagnosis of different types

Establish associated lesions cardiac and extracardiac

Counselling can be challenging in some cases as management strategy may be limited or unclear

Management and outcome influenced by extent of associated anomalies both cardiac and extracardiac

Delivery

- Balanced AVSD which is isolated and no AVVR – local delivery and postnatal review
- All others consider delivery at tertiary centre or early referral to tertiary centre

Associations

Data from Evelina London Children's Hospital/ Guy's & St Thomas' NHS Foundation Trust

Huggon et al 2000 JACC n=320

- 40% of AVSD had T21
- 10% other chromosomal abn
- 12% had RAI
- 20% had LAI
- 13% had ECA

Sharland 2012 - Fetal Cardiology Simplified n=687

- 36% AVSD chromosomal abnormality
  - 86% of these trisomy 21
- 24% Isomerism
  - 16% LAI
  - 8% RAI
- 4% other ECA

Improvements in screening have led to more isolated cases in later part of series