Prenatal Evaluation of the Fetus with Atrioventricular Canal Defect

Children's Mercy Fetal Cardiology Education Series

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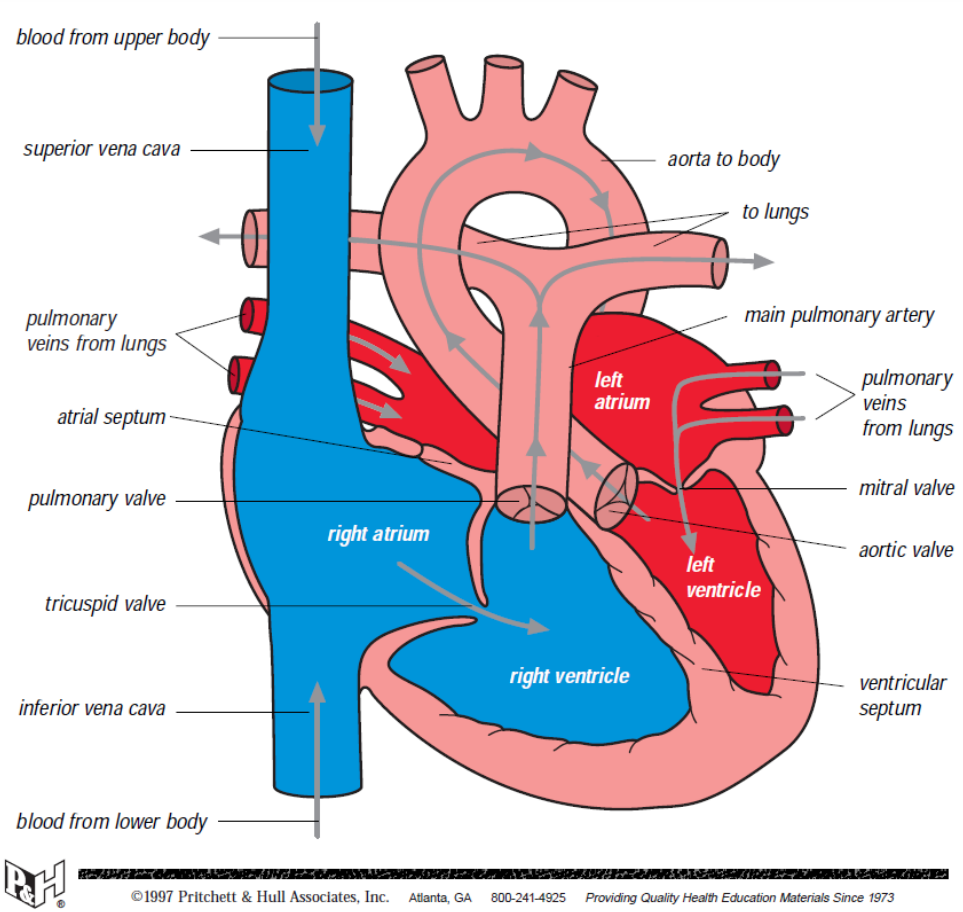
Disclosures

- No disclosures

Objectives

- Understand the basic anatomy and embryologic basis of atrioventricular canal/septal defects (AVSDs)
- Define fetal imaging goals for AVSDs
- Recognize aspects of AVSD anatomy on shared AVSD fetal imaging cases

Normal Heart



Atrioventricular Septal Defect

- AKA AV canal defect, endocardial cushion defect
- 40-45% with trisomy 21/Down syndrome have CHD: ~40% of these have AVSD
- Heterotaxy
- Embryologically: failure of the endocardial cushions to fuse creating a defect in the AV septum
- Fetal diagnosis feasible and allows for counseling and identification of noncardiac defects (trisomy 21)

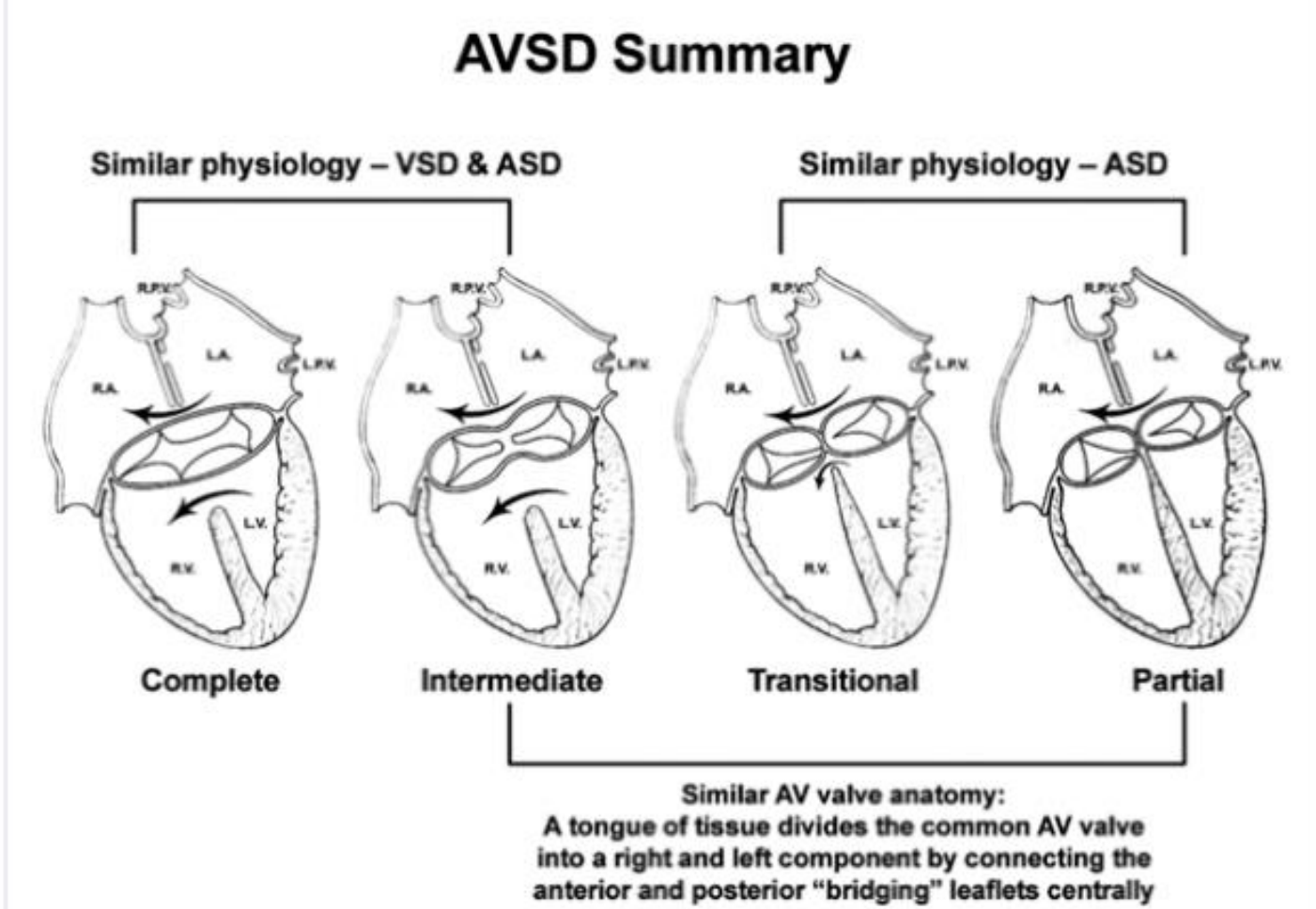
Atrioventricular Septal Defect

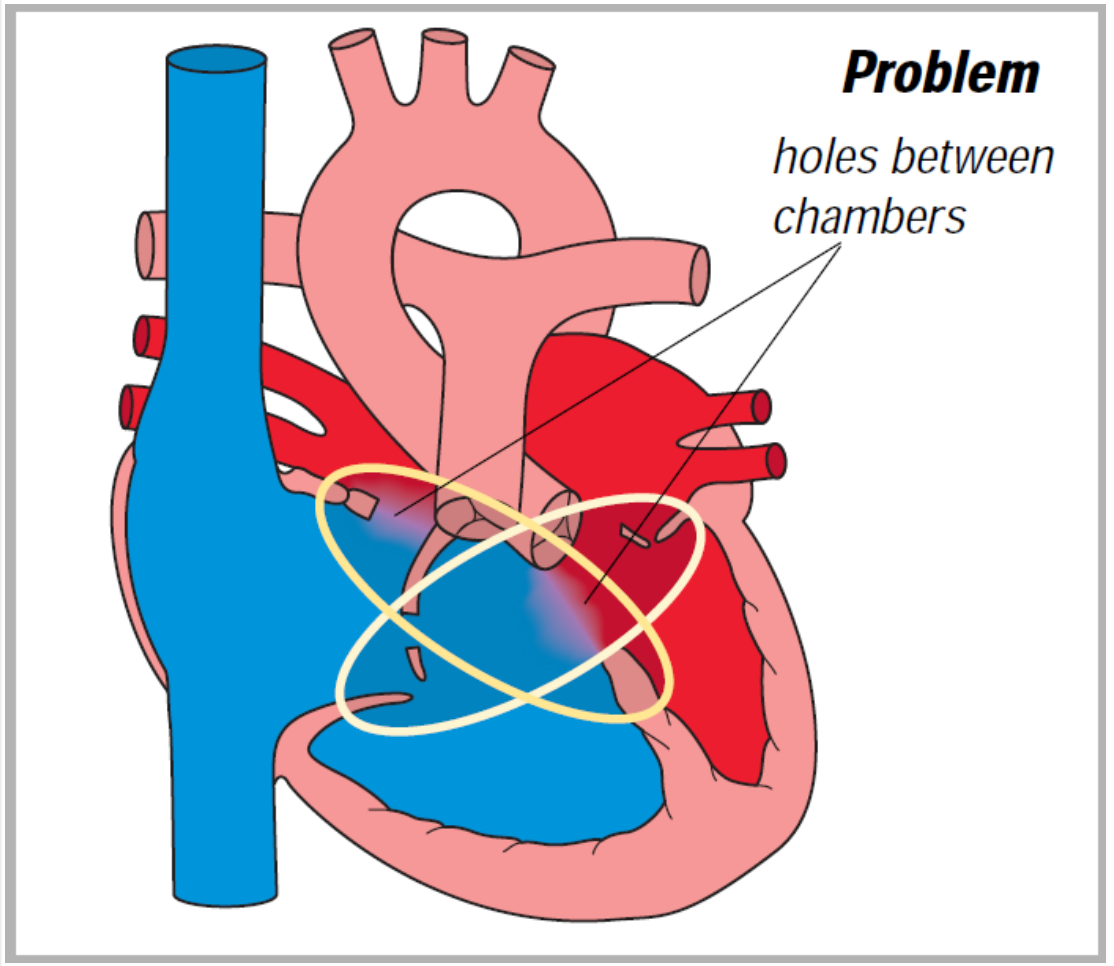
- Absence of the atrioventricular (AV) portion of the septum
  - Exclusive atrial communication
  - Combined atrial septal defect and ventricular septal defect
  - Exclusive ventricular communication
- Maldevelopment of AV valves resulting in common inlet to both ventricles
- Common AV valve is abnormal, and incompetence (regurgitation) frequently occurs

Atrioventricular Septal Defect

- Complete versus partial AVSD
- Balanced versus unbalanced (i.e. ventricular dominance, single ventricle)
- Complete form:
  - Primum atrial septal defect
  - Inlet ventricular septal defect
  - Single common AV valve annulus/common AV valve junction
- Partial form: primum ASD and cleft mitral valve

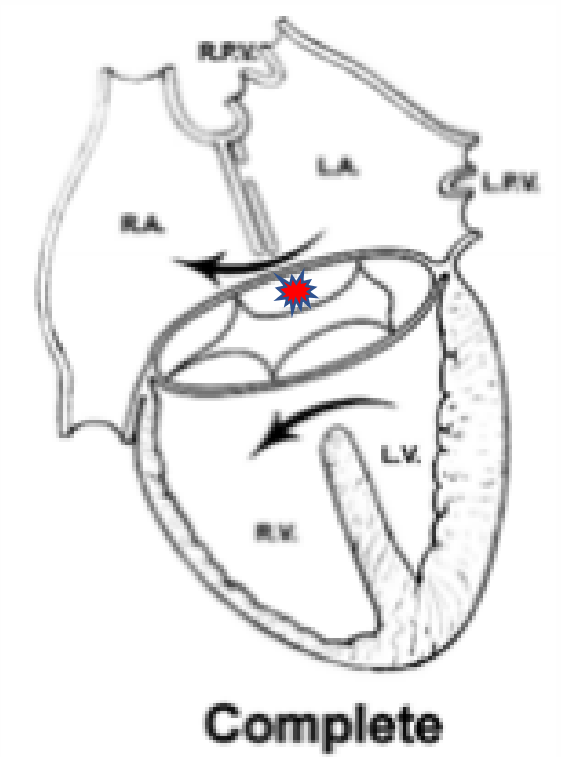
Anatomy





Allen, Hugh D., et al. *Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. 2013.

Common AV Valve Anatomy

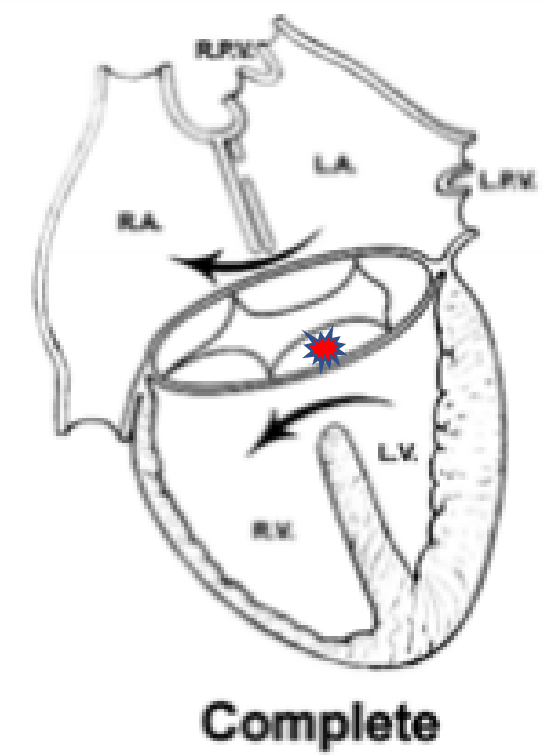


Complete

- Superior bridging leaflet

Allen, Hugh D., et al. *Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. 2013.

Common AV Valve Anatomy

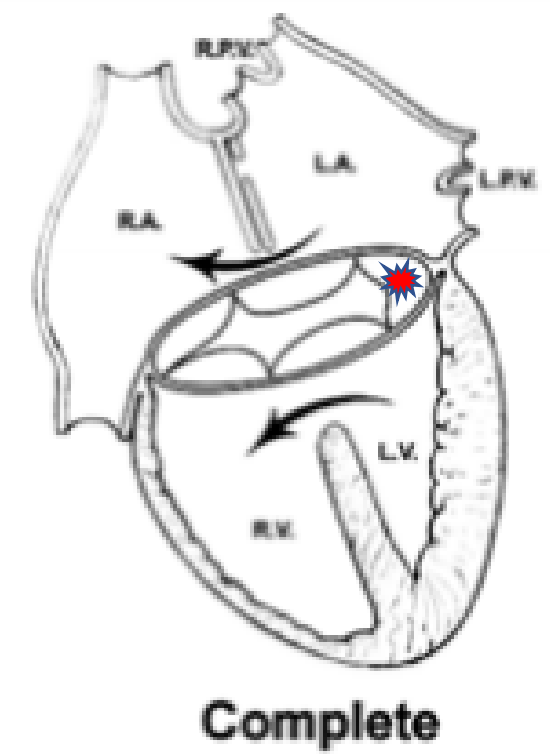


Complete

- Inferior bridging leaflet

Allen, Hugh D., et al. *Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. 2013.

Common AV Valve Anatomy

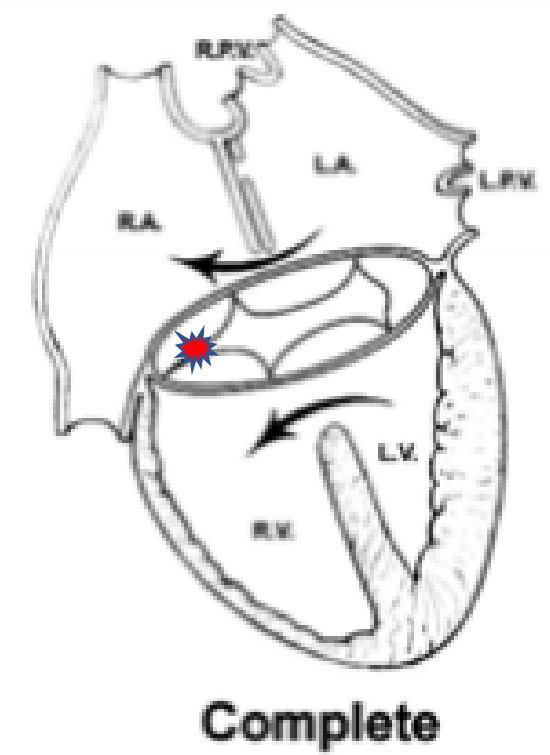


Complete

- Left mural leaflet

Allen, Hugh D., et al. *Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. 2013.

Common AV Valve Anatomy



Complete

- Right anterior and posterior leaflets

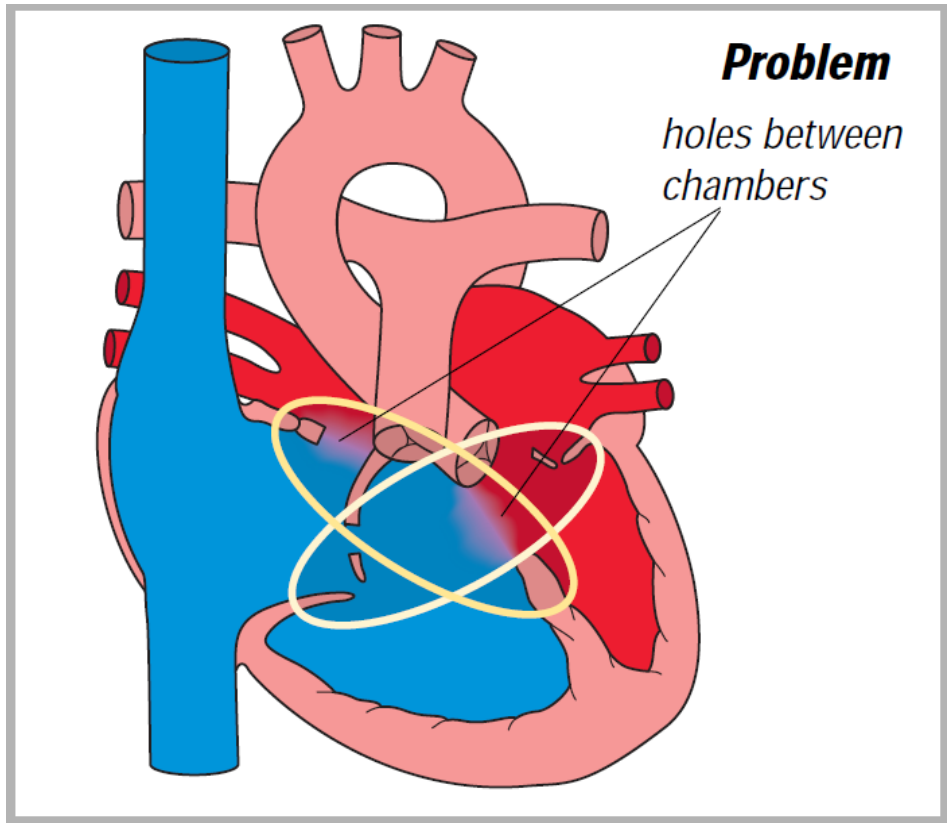
Allen, Hugh D., et al. *Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. 2013.

Common AV Valve Anatomy

- Rastelli Classification (superior bridging leaflet)
  - Type A: superior bridging leaflet divided at ventricular septum and attaches to septum
  - Type B: rare, attaches to superior RV papillary muscle
  - Type C: free floating, no attachments to septum

Fetal Physiology

- Little impact on fetal circulation unless significant common AV valve regurgitation
- Pulmonary vascular resistance in utero is elevated
  - Little left to right shunting → no significant volume load
- Complete heart block



Associated Lesions

- Patent ductus arteriosus
- Secundum atrial septal defect
- Coarctation of the aorta
- Additional VSDs
- Tetralogy of Fallot (almost exclusively seen in patients with trisomy 21)

AVSD Fetal Echo Features

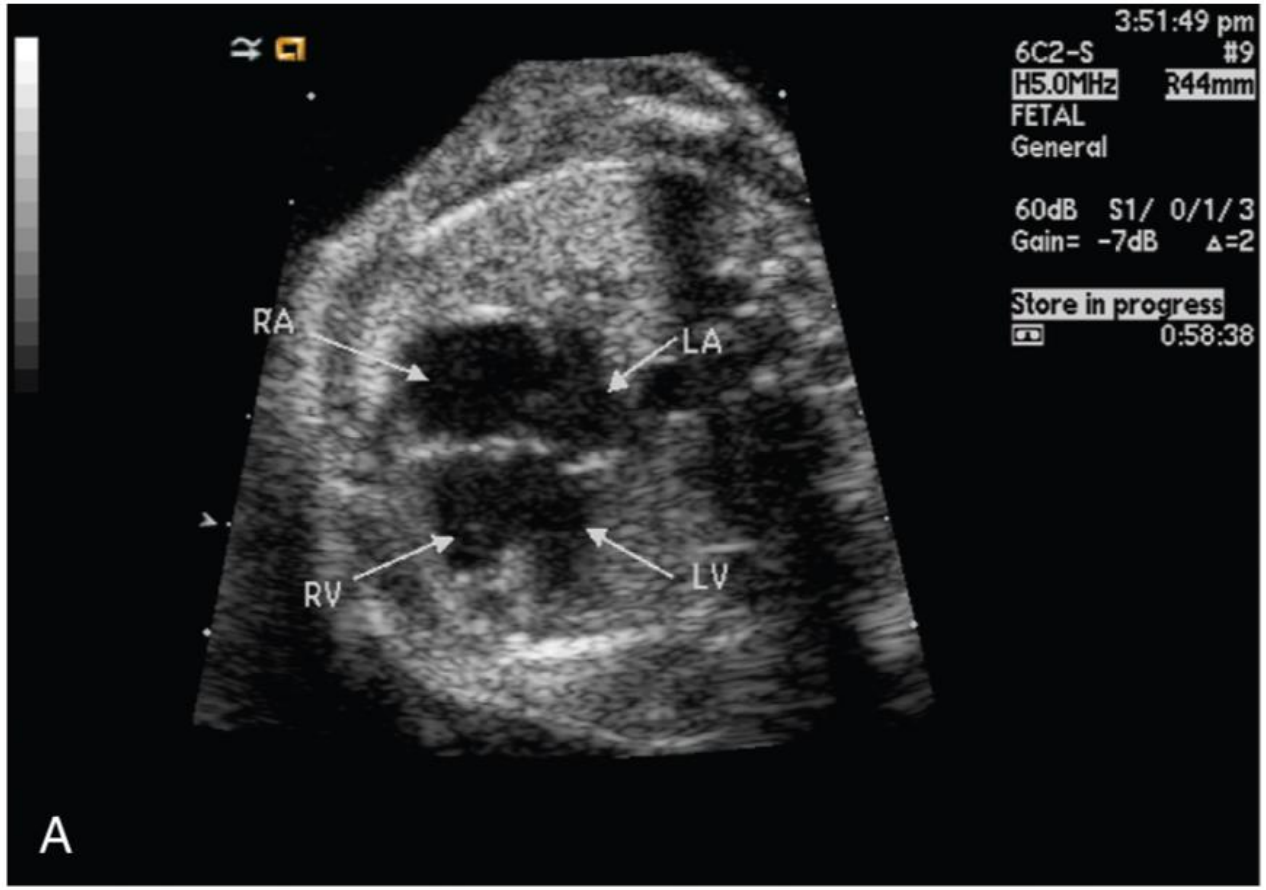
Key Echocardiographic Features

- Size of the atrial septal defect
- Size of the ventricular septal defect (large = complete, small = transitional, none = incomplete)
- Valve attachments to the crest of the septum
- Presence or absence and degree of atrioventricular valve regurgitation
- Degree of balance or unbalance of the atrioventricular valve over the two ventricles
- Relative size of the ventricular cavities
- Presence or absence of left ventricular outflow tract obstruction (valve tissue obstructing or narrowed pathway due to small left ventricular outflow tract)

Rychik, Jack. Zhiyun, Tian. *Fetal Cardiovascular Imaging: A Disease-Based Approach.* Elsevier Saunders, 2012.

AVSD Fetal Echo Features

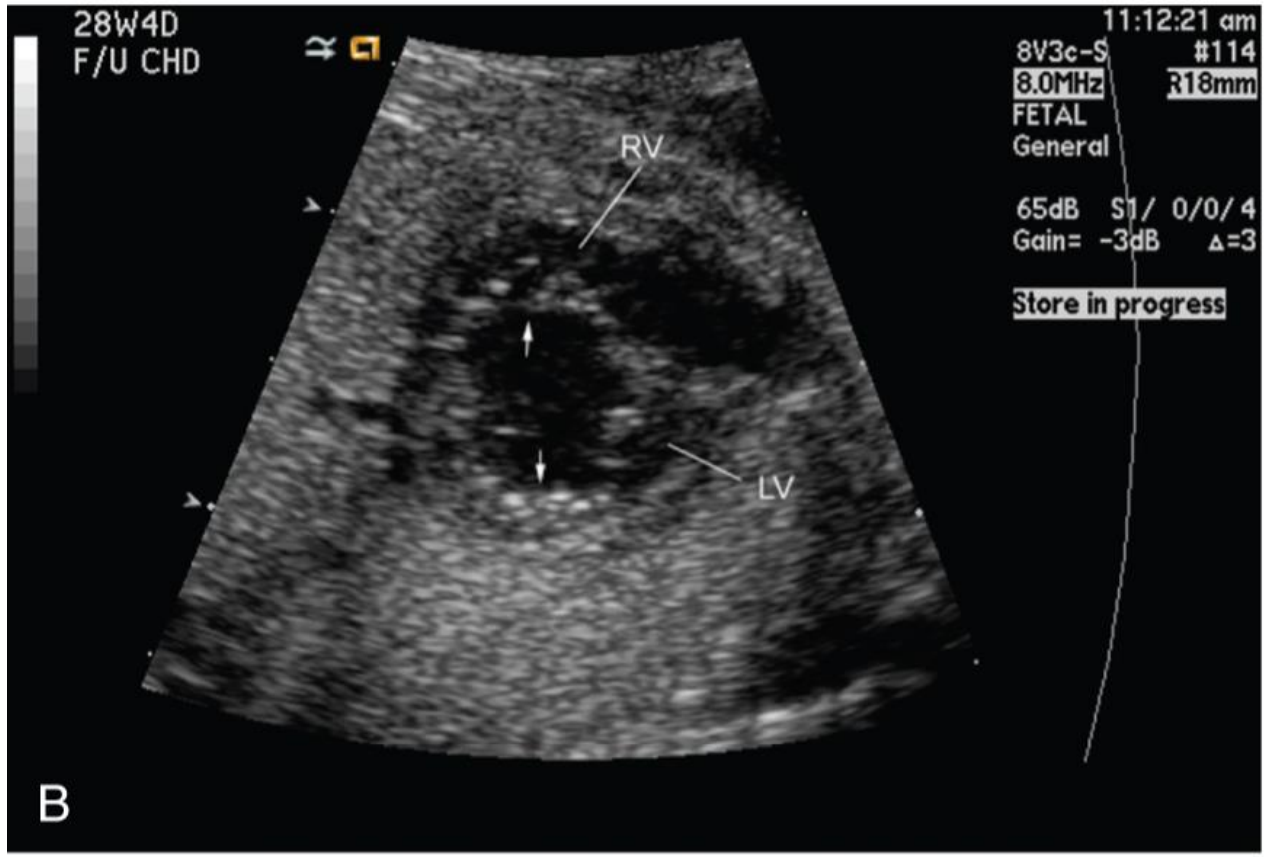
- Four-chamber view identifies diagnosis



Rychik, Jack. Zhiyun, Tian. *Fetal Cardiovascular Imaging: A Disease-Based Approach.* Elsevier Saunders, 2012.

AVSD Fetal Echo Features

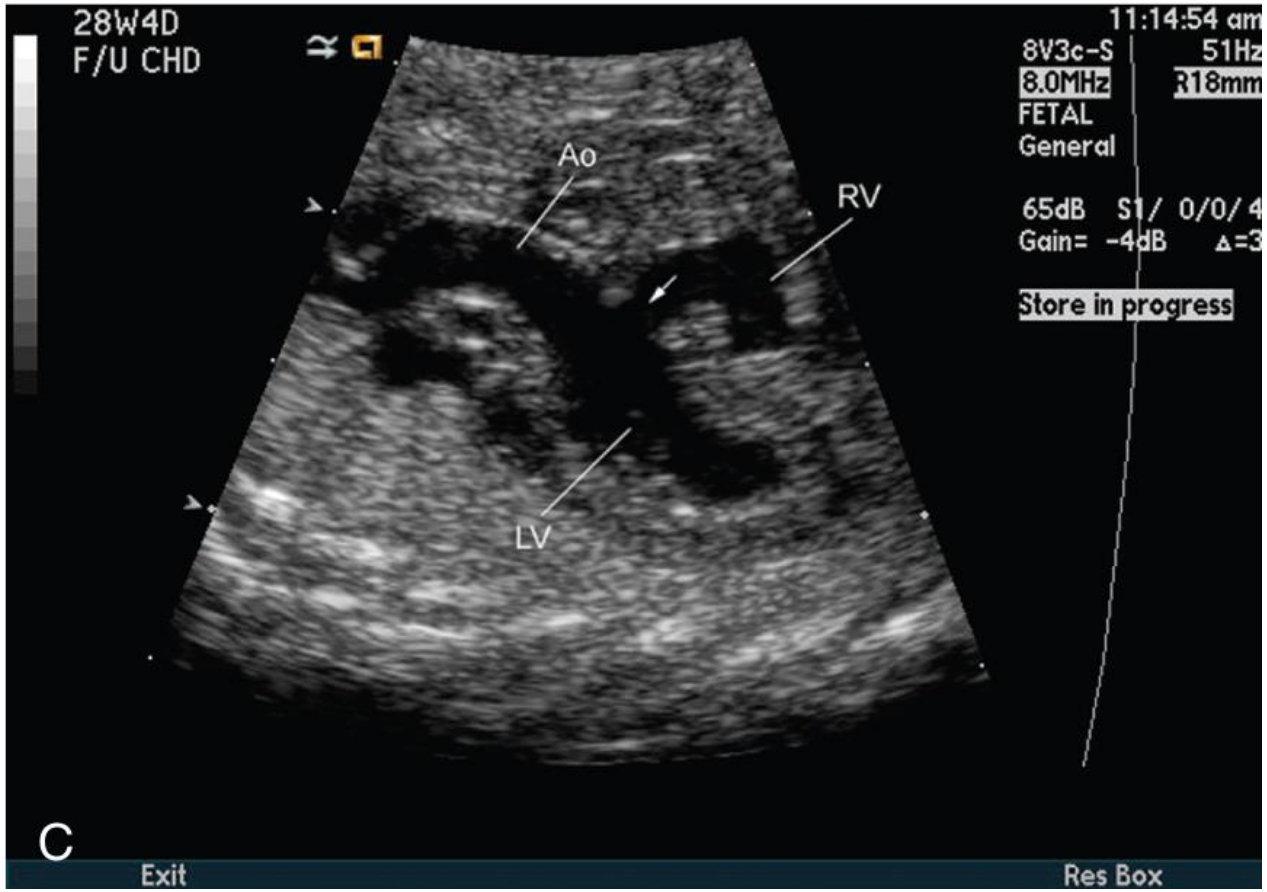
- Short-axis view: common AV valve en face with superior and inferior bridging leaflets crossing into both ventricles



Rychik, Jack. Zhiyun, Tian. *Fetal Cardiovascular Imaging: A Disease-Based Approach.* Elsevier Saunders, 2012.

AVSD Fetal Echo Features

- Long-axis imaging of LV to aorta demonstrates elongated, scooped out or gooseneck LV outflow tract



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Gooseneck Deformity in AVSD

Figure 29.14 Gooseneck left ventricular outflow tract (LVOT) deformity in AVSD. Because of the anterior displacement of the LVOT in AVSD, the elongated LVOT has been described as a "gooseneck" with echocardiographic (**left**) and angiographic (**right**) imaging. **Center:** Pediatric and adult goosenecks in Minnesota.

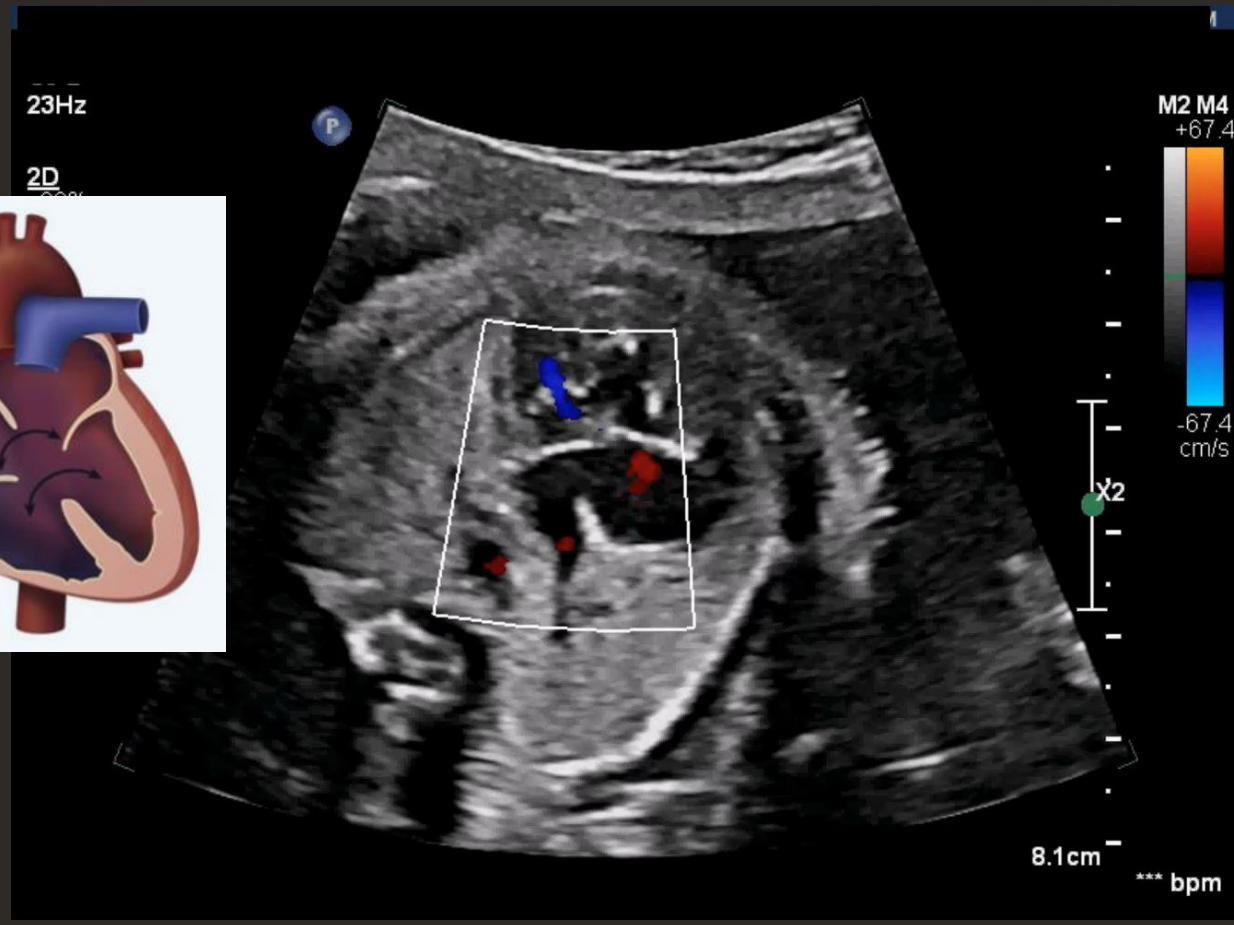
AVSD Fetal Echo Features

- Color Doppler identifies fetal AV valve regurgitation
  - If more than mild in utero → structural abnormality of the valve
  - May worsen after birth due to change in loading conditions
- Fetal rhythm evaluation: complete heart block sometimes seen (poor prognosis)

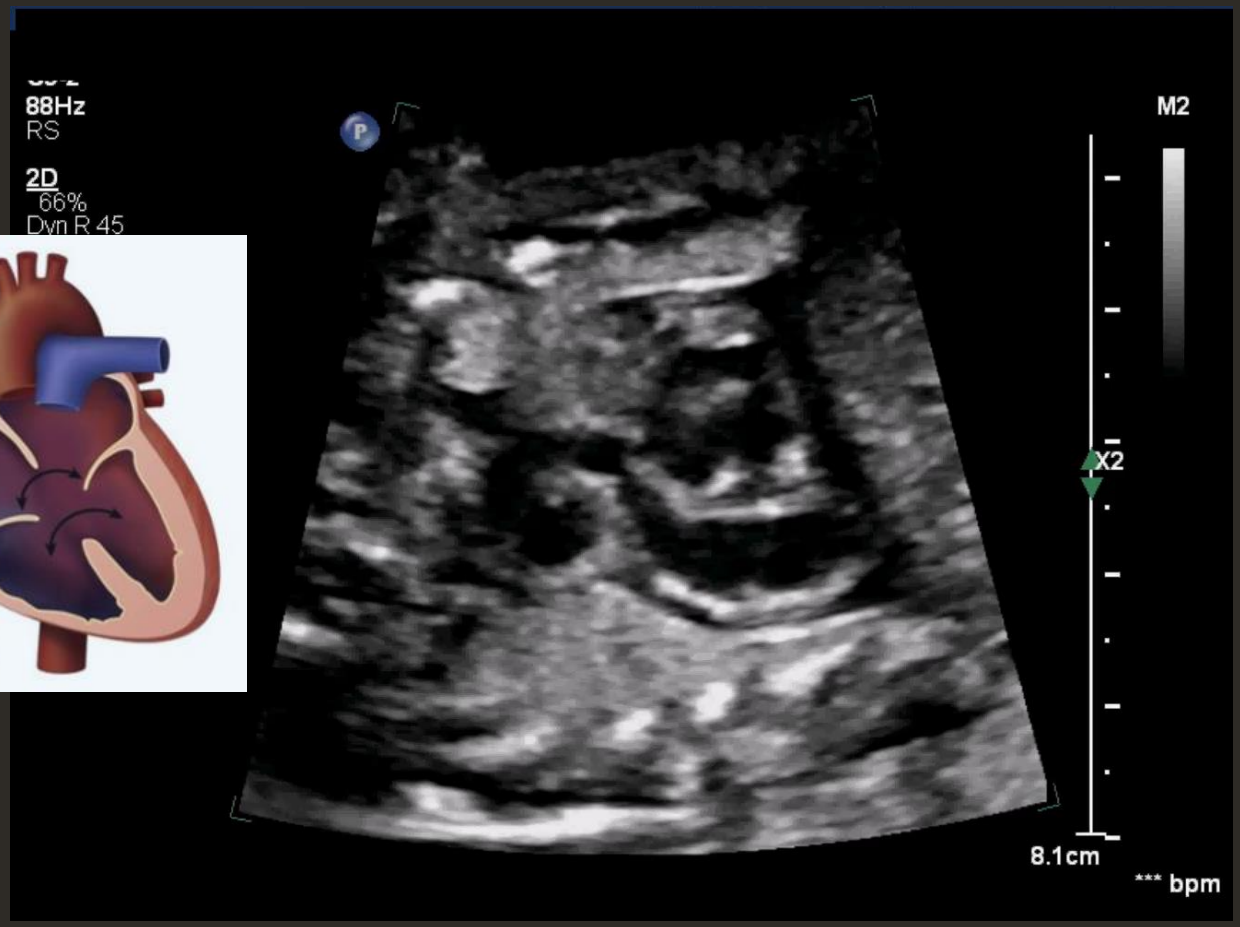
Degree of “Balance” of the Common AV Valve

- Normally sits approximately 60% over RV
- If >60% over RV → RV dominant unbalanced AVSD
  - If RV dominant: evaluate left sided structure sizes
- Equal sitting of the common AV valve over both ventricles or >50% over LV → LV dominant unbalanced AVSD
  - If LV dominant: evaluate for RV hypoplasia, pulmonary stenosis, branch pulmonary artery narrowing

Complete, balanced AVSD

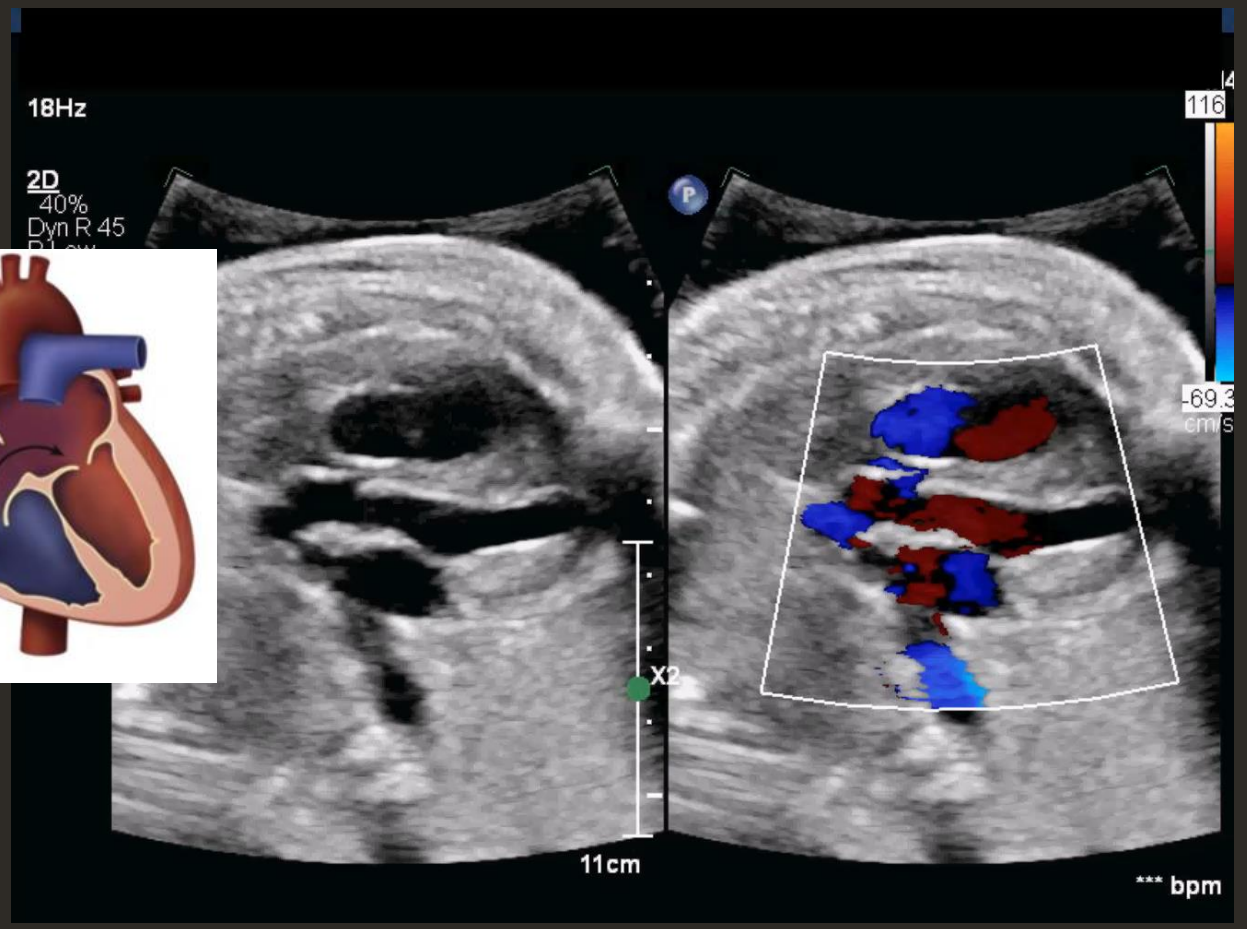


Complete, balanced AVSD



Complete, balanced AVSD: AV valve en face

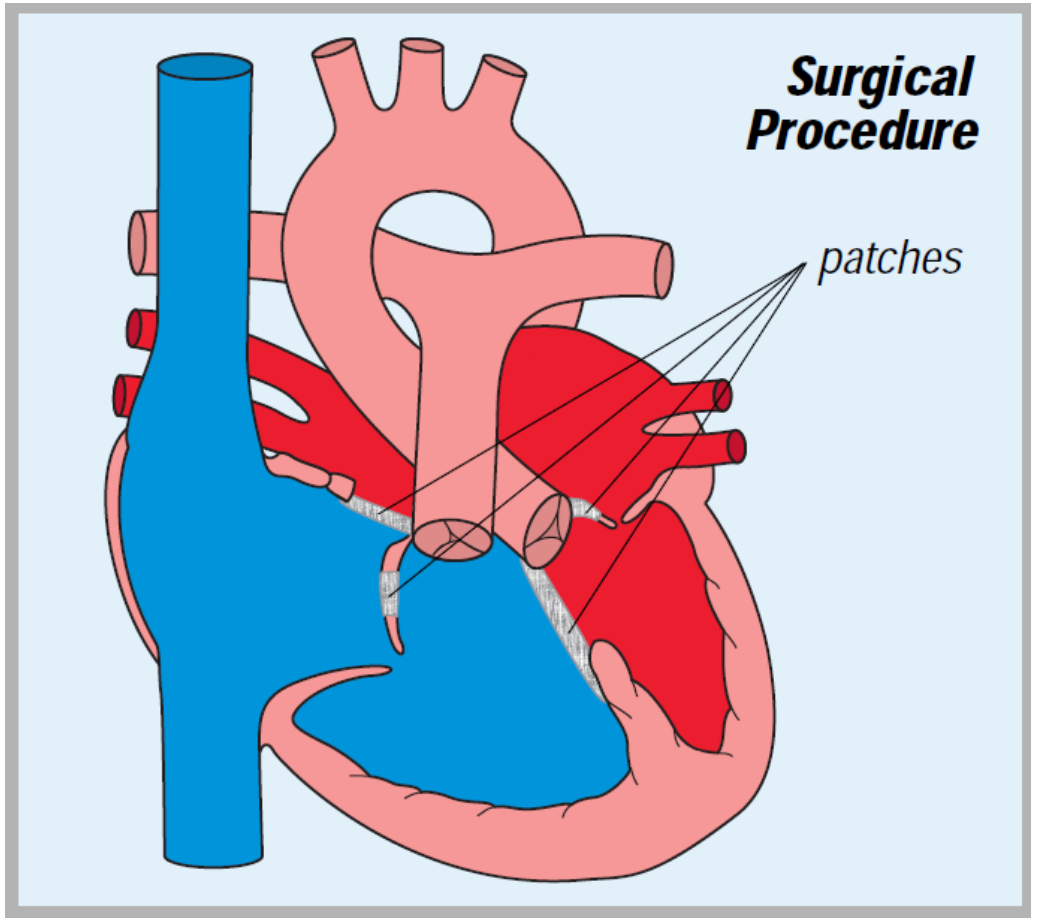
Partial/Incomplete AVSD



Unbalanced AVSD

AVSD Surgical Repair and Outcomes

- Complete, balanced AVSD
  - Surgical repair around 4-6 months of age
  - Long-term survival excellent
  - Residual lesions: valve regurgitation, valve stenosis, subaortic stenosis
- Unbalanced AVSD
  - Single ventricle palliation



Thank you!

References

1. Allen, Hugh D., et al. *Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. Wolters Kluwer Health, Lippincott Williams & Wilkins, 2013.
2. Park, Myung K., *Park's Pediatric Cardiology for Practitioners*. Mosby, 2014
3. Rychik, Jack. Zhiyun, Tian. *Fetal Cardiovascular Imaging: A Disease-Based Approach*. Elsevier Saunders, 2012.

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