Hypoplastic Left Heart Syndrome (HLHS)

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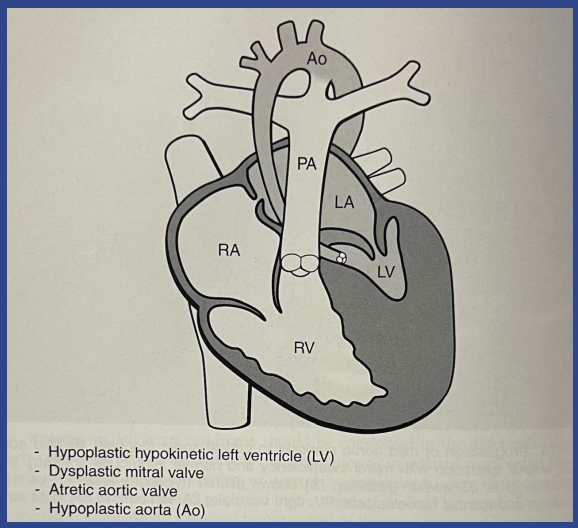
Objectives

- Review:
  - Definitions
  - Epidemiology
  - Anatomy
- Discuss Embryology
- Review key measurements to obtain on Fetal Echo



Abuhamad, A., & Chaoui, R. (2010). *A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts*. Lippincott Williams & Wilkins.

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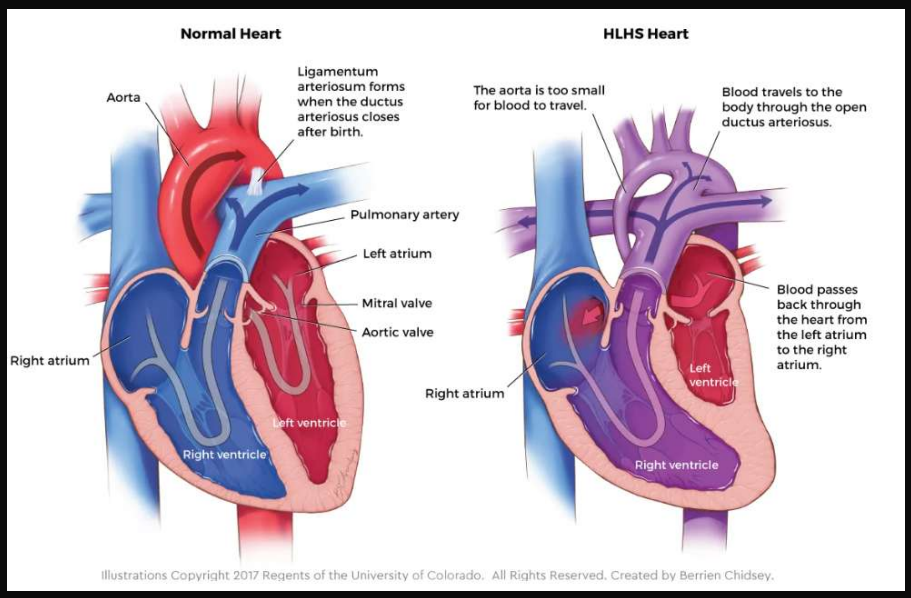

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History of Hypoplastic Left Heart Syndrome

- First published cases of aortic atresia were reported by Romberg in 1846 and Canton in 1849 and are more or less restricted to brief pathologic and anatomic descriptions.
- 1851 the account of Dr. Bardeleben was probably the first complete description of the clinical features, the pathologic anatomic characteristics and the pathophysiology of the hypoplastic left- heart syndrome.

Gehrmann, Josef et al. Hypoplastic Left-Heart Syndrome, CHEST, Volume 120, Issue 4, 1368 - 137

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History of Hypoplastic Left Heart Syndrome

- 1851 German pathologist Barbedelan reported the autopsy finding of a 6-month-old infant who died of severe asphyxia.
- Evaluation of the cardiac anatomy revealed hypoplasia of the left ventricular cavity without communication to the aorta, severe hypoplasia of the ascending aorta, and a ductus arteriosus.
- In 1851 Barbedelan hypothesized that the systemic circulation was dependent on patency of the ductus arteriosus.

Hypoplastic Left-Heart Syndrome: The First Description of the Pathophysiology in 1851; Translation of a Publication by Dr. Bardeleben From Giessen, Germany - ScienceDirect

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History of Hypoplastic Left Heart Syndrome

- 1958 Noonan and Nadas coined the term hypoplastic left heart syndrome (HLHS)
- Described a constellation of malformation resulting from severe underdevelopment of any left-sided cardiac structures.

Mohanty SR, Patel A, Kundan S, Radhakrishnan HB, Rao SG. Hypoplastic left heart syndrome: current modalities of treatment and outcomes. *Indian J Thorac Cardiovasc Surg*. 2021;37(Suppl 1):26-35. doi:10.1007/s12055-019-00919-7

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Defining Characteristics

- Collection of heterogeneous congenital heart anomalies in which the left ventricle, by virtue of structural abnormalities, is incapable of providing adequate systemic perfusion.
- May include abnormalities such as:
  - Mitral stenosis or atresia
  - Left ventricular hypoplasia
  - Aortic valve stenosis or atresia
  - Ascending aorta hypoplasia

Lai, Wyman W., et al., editors. *Echocardiography in Pediatric and Congenital Heart Disease: From Fetus to Adult*. Third edition, Wiley-Blackwell, 2021

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Incidence

- Birth incidence of HLHS is 0.1 to 0.25 per 1000 live births
- HLSH accounts for 3.8% of all congenital cardiac abnormalities and up to seven-tenths of cases occur in boys
- Although HLHS is one of the most commonly diagnosed congenital heart abnormalities in utero, it is still missed in a significant proportion of fetuses.

Abuhamad, A., & Chaoui, R. (2010). *A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts*. Lippincott Williams & Wilkins

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Genetics

- Turner syndrome (chromosomes 45, XO),
  - Should be investigated in every female infant with HLHS
- Trisomy 18
- 22q11.2 deletion
- Jacobsen
- Kabuki
- Rubinstein-Taybi
- Smith-Lemli-Opitz
- Adams-Oliver
- Beckwith-Wiedemann
- Meckel-Gruber
- VACTERL

Lai, Wyman W., et al., editors. *Echocardiography in Pediatric and Congenital Heart Disease: From Fetus to Adult*. Third edition, Wiley-Blackwell, 2021.

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Genetics

- Strong familial clustering of HLHS
- Recurrence risk of 21% in children of an affected parent
- Risk of 8% if a sibling is affected

Lai, Wyman W., et al., editors. *Echocardiography in Pediatric and Congenital Heart Disease: From Fetus to Adult*. Third edition, Wiley-Blackwell, 2021

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Neurologic Impact

- Increased risk of congenital brain abnormalities
  - Decreased cortical volumes
  - Agenesis of the corpus callosum
  - Holoprosencephaly
  - Cortical mantle malformation
- Increased risk of developing acquired injury such as hypoxic-ischemic lesions and intracranial hemorrhage
- High risk for neurodevelopmental impairment

Lai, Wyman W., et al., editors. *Echocardiography in Pediatric and Congenital Heart Disease: From Fetus to Adult*. Third edition, Wiley-Blackwell, 2021

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Types

- Three types based on the size and function of the mitral and aortic valves

- Mitral and aortic stenosis (MS/AS)
- Mitral stenosis and aortic atresia (MS/AA)
- Mitral and aortic atresia (MA/AA)

Lai, Wyman W., et al., editors. *Echocardiography in Pediatric and Congenital Heart Disease: From Fetus to Adult*. Third edition, Wiley-Blackwell, 2021

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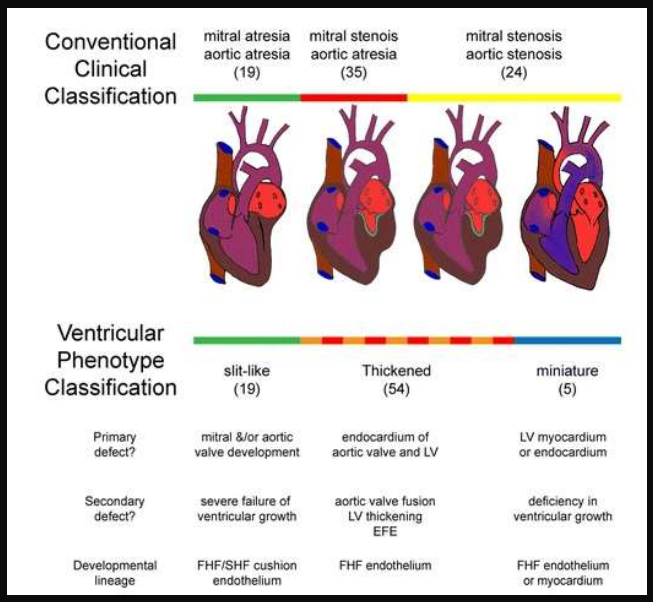

Crucean, A., Alqahtani, A., Barron, D.J. *et al.* Re-evaluation of hypoplastic left heart syndrome from a developmental and morphological perspective. *Orphanet J Rare Dis* **12**, 138 (2017). https://doi.org/10.1186/s13023-017-0683-4

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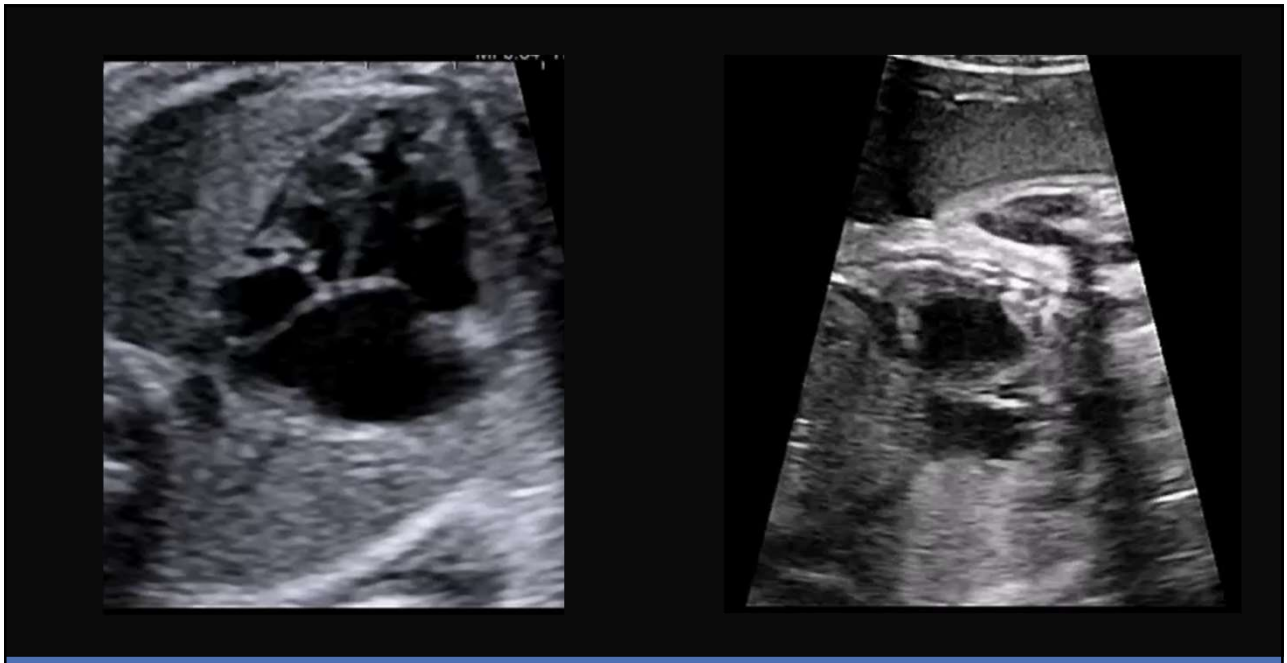

HLHS (MS/AA)

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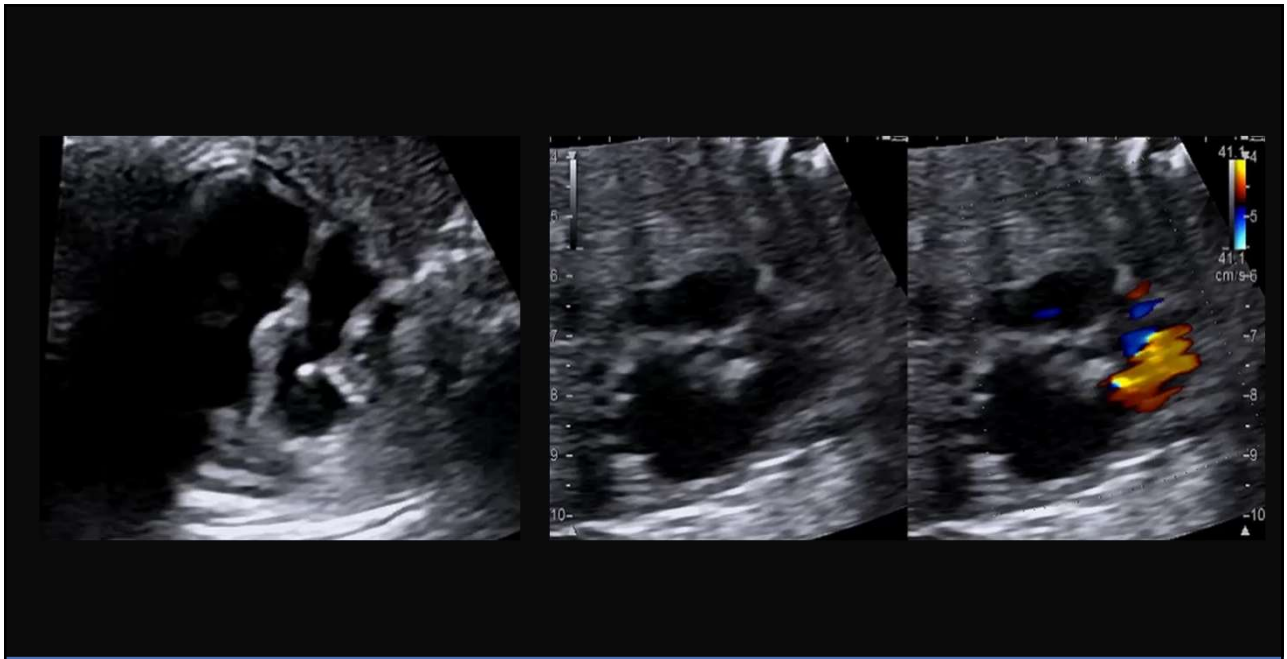

HLHS (MS/AS)

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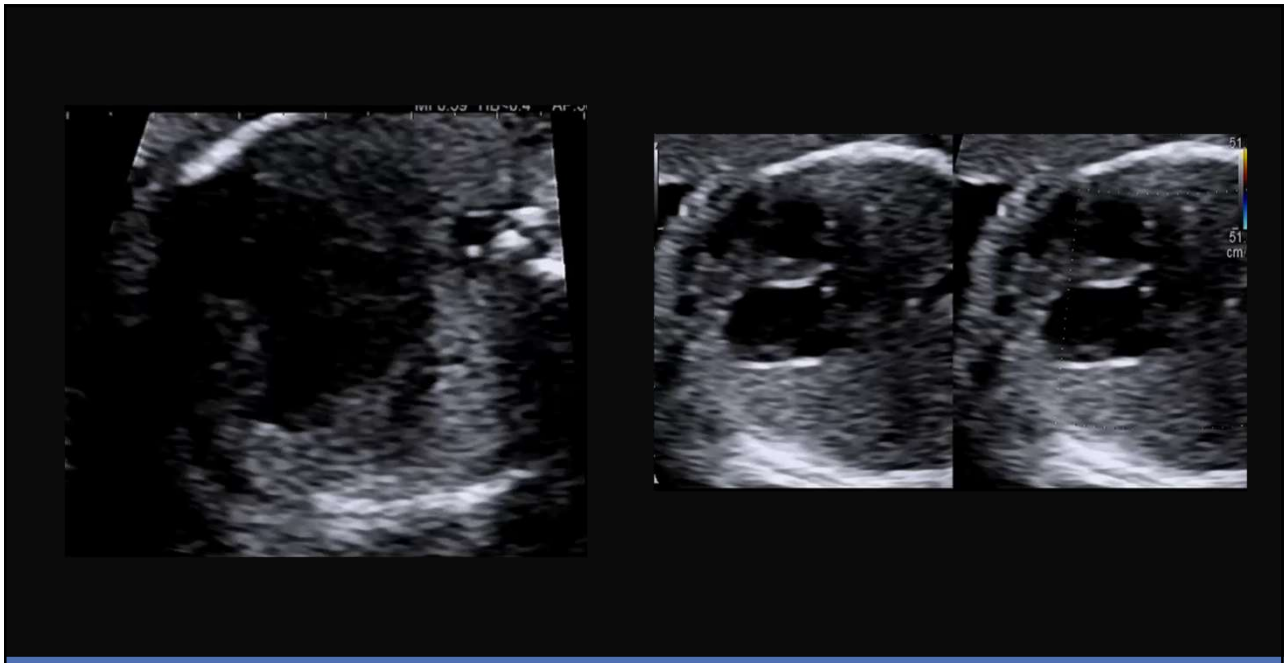

HLHS (MA/AA)

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Early Gestational Imaging

- Detectable in early gestation at 11-14 weeks
  - Primarily in cases with a combined MA/AA, showing an absence of severely hypoplastic left ventricle
- Also, can develop between the first and second trimesters
  - emphasizing that a normal-appearing four-chamber view at the time of nuchal translucency measurement does not rule out the development of HLHS in later gestation.

Abuhamad, A., & Chaoui, R. (2010). *A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts*. Lippincott Williams & Wilkins

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Ultrasound Findings

- 4 chamber is remarkably small, hypokinetic
  - This can vary and be absent, small of normal size or even dilated, but in all cases this ventricle is hypocontractile with absent function.
- Apex of the heart is predominantly formed by the right ventricle
- Bright echogenic inner wall due to the associated endocardial fibroelastosis (EFE)

Abuhamad, A., & Chaoui, R. (2010). *A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts*. Lippincott Williams & Wilkins

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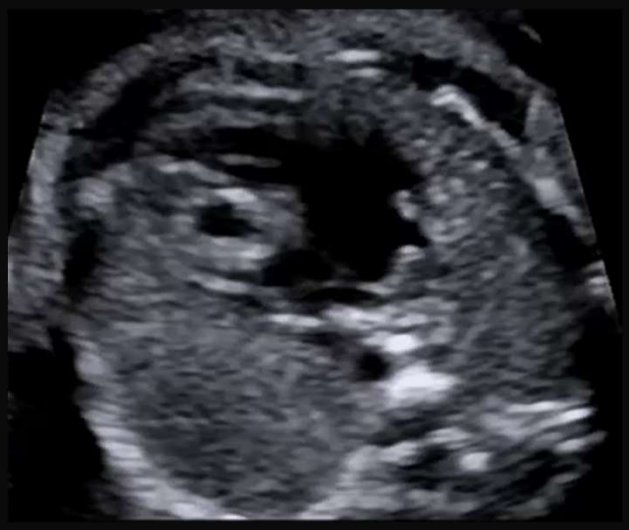

2D

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Color Doppler Findings

- Minimal to absent filling of the left ventricle
- In cases with a patent mitral valve, mitral regurgitation can be found.
- Left to right shunt across the foramen ovale is found, due to increased pressure in the left atrium
- Lack of forward flow across the atretic aortic valve
- 3VV shows reversal of flow across the aortic isthmus and transverse aortic arch

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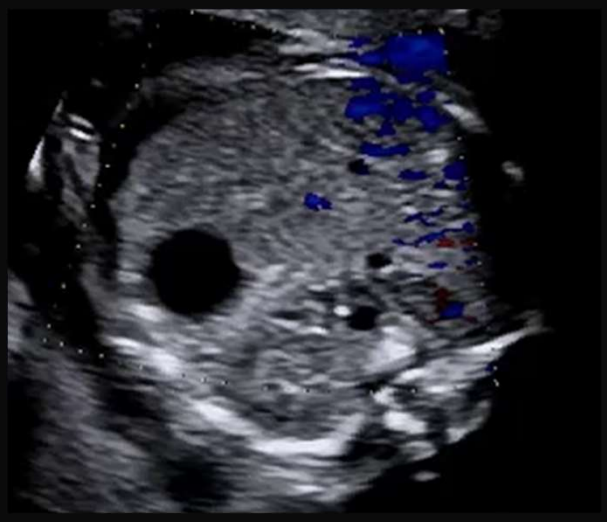

COLOR

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Atrial Septum and Pulmonary Vein Dopplers

- Magnitude of atrial level restriction is assessed by direct inspection of the atrial septum on 2D as well as color Doppler.
- Pulsed Doppler interrogation of the pulmonary veins is important for assessing the degree of reversal of pulmonary venous flow with atrial contraction
  - The greater is the degree of impediment to left atrial egress.
- Patency of the interatrial communication is critical for normal lung development
  - as increased left atrial pressure in this setting is transmitted back to the pulmonary vasculature, leading to pulmonary vein dilation and smooth muscle proliferation in the walls of the pulmonary veins.

Lai, Wyman W., et al., editors. *Echocardiography in Pediatric and Congenital Heart Disease: From Fetus to Adult*. Third edition, Wiley-Blackwell, 2021

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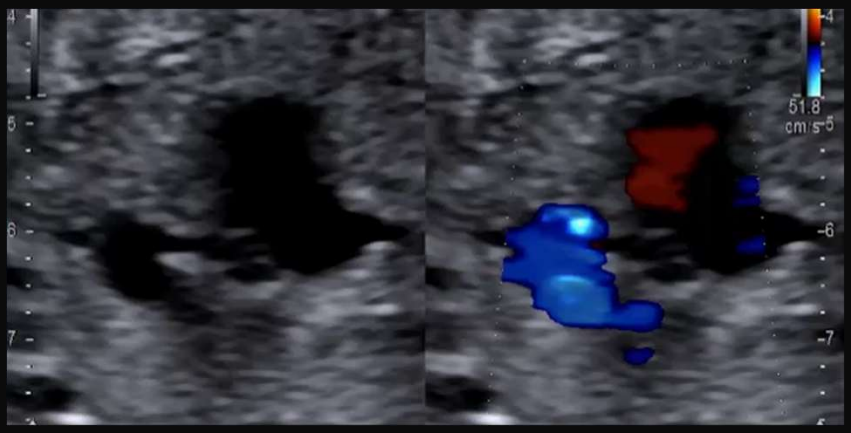

RESTICTIVE ATRIAL SEPTUM

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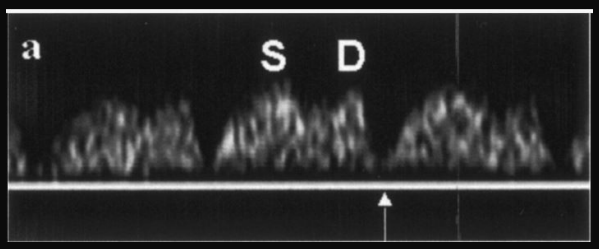
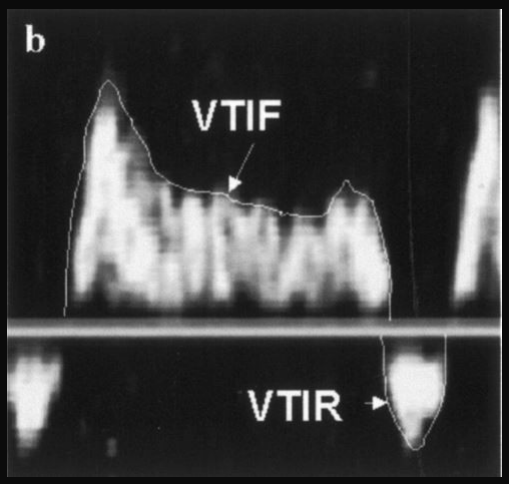
Pulmonary Vein Doppler





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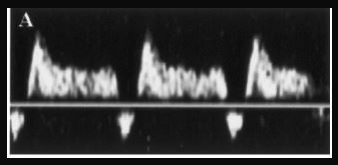
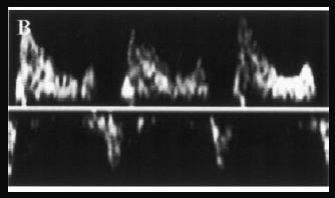
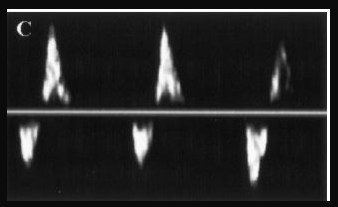
Pulmonary Vein Doppler







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Prognosis

- Serial prenatal ultrasound evaluation every 4-6 weeks is recommended to assess
  - Fetal growth
  - Tricuspid valve function
  - Flow across the foramen ovale
  - Pulmonary vein Doppler patterns
- Presence of tricuspid valve dysfunction and/or restriction across the foramen ovale case a poor prognostic outlook.

Abuhamad, A., & Chaoui, R. (2010). *A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts*. Lippincott Williams & Wilkins

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CASE #1

26 year old referred for prenatal diagnosis in Mexico of Hypoplastic Left Heart Syndrome

Physicians in Mexico gave a 1% chance of survival.

Mom obtained a medical visa for 1 year and arrived at labor and delivery unit with abdominal pain

33w1d gestation

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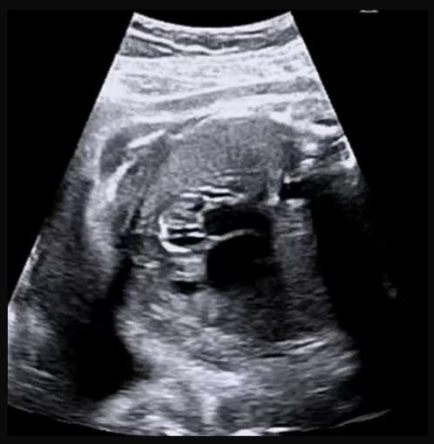
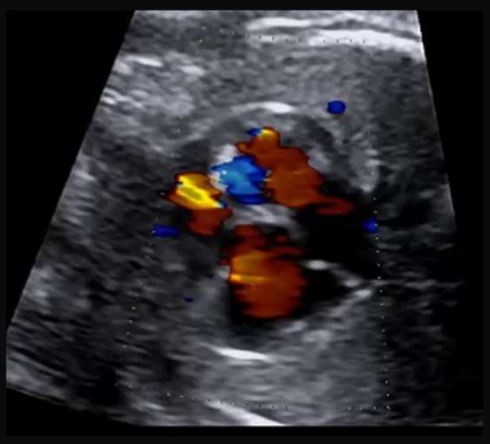
33W1D





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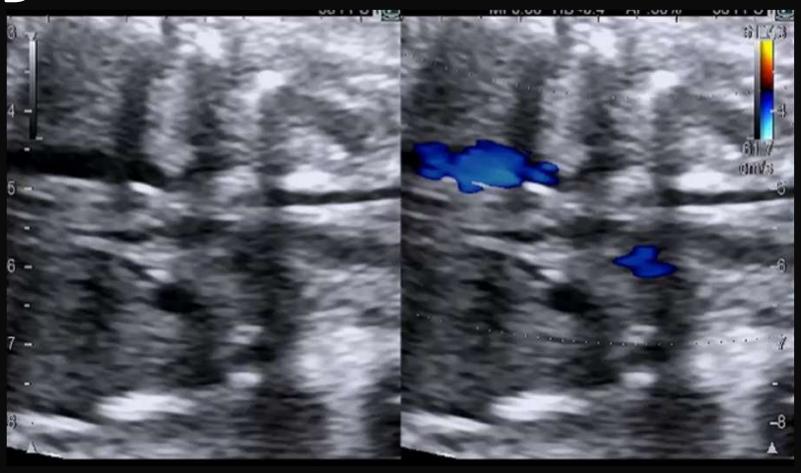
33W1D



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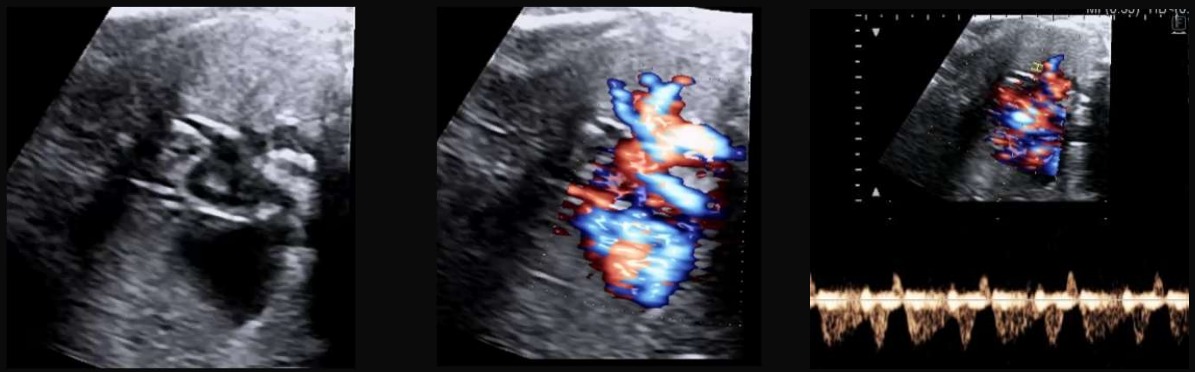
33W1D



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Three weeks later ☺

Delivered at 36weeks 2days

Placed on nasal cpap shortly after birth.

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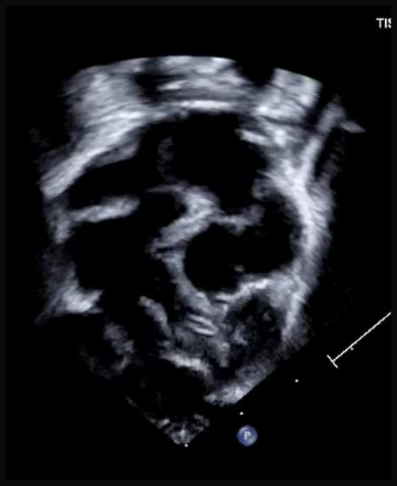
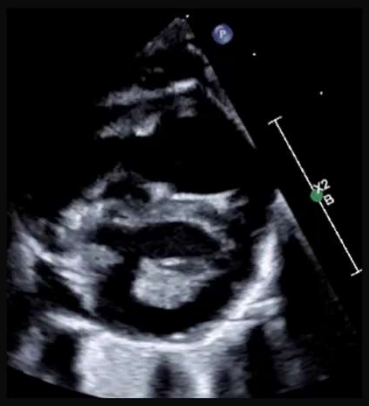
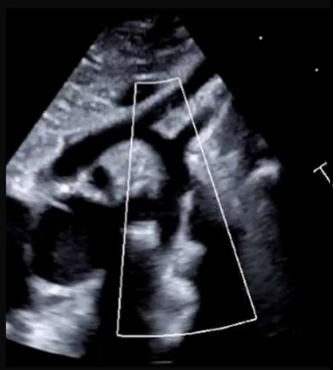
Post natal







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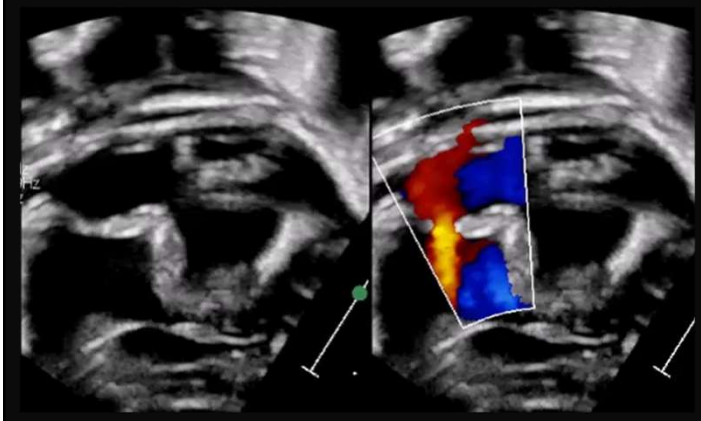
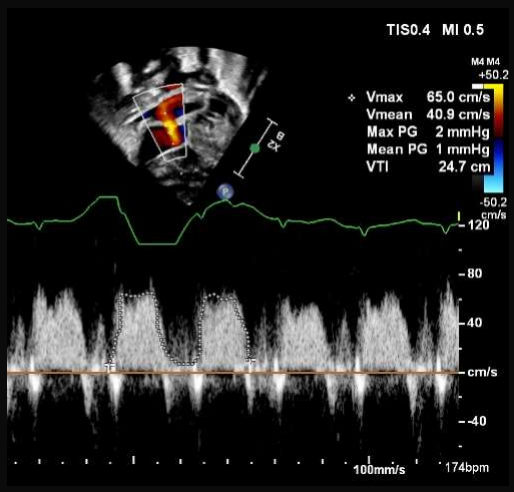
Atrial Septum At Birth





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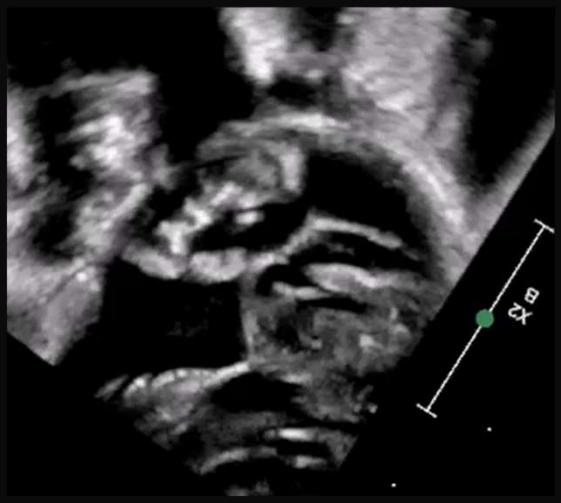
Post Natal Balloon Septostomy



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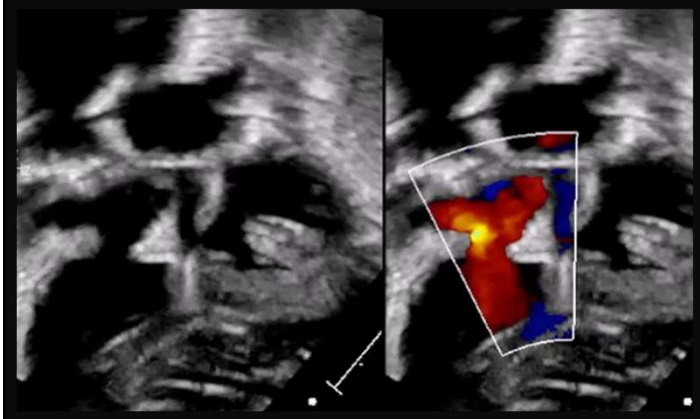
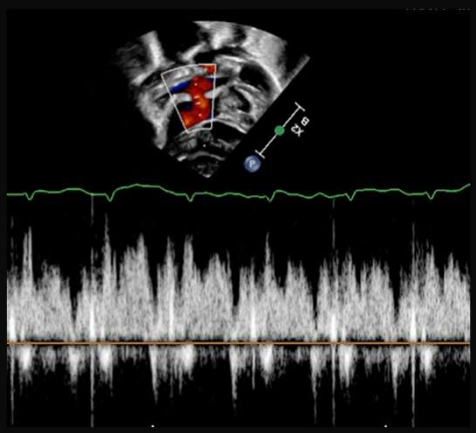
Atrial Septum Post Balloon Septostomy





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CASE #2

- 27 Year old female
- 24W6D gestation
- Referred for an abnormal OB ultrasound concerning for Hypoplastic Left Heart Syndrome

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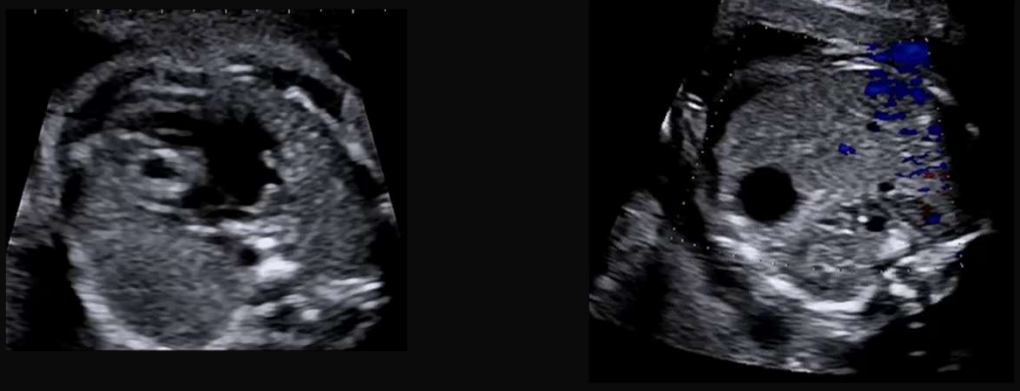
24W6D



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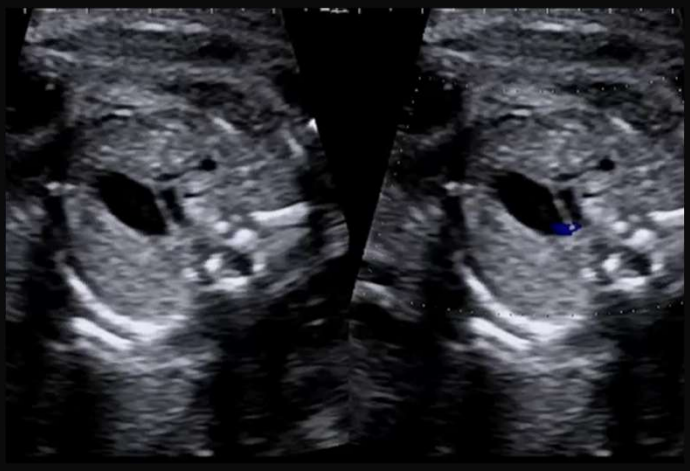
24W6D



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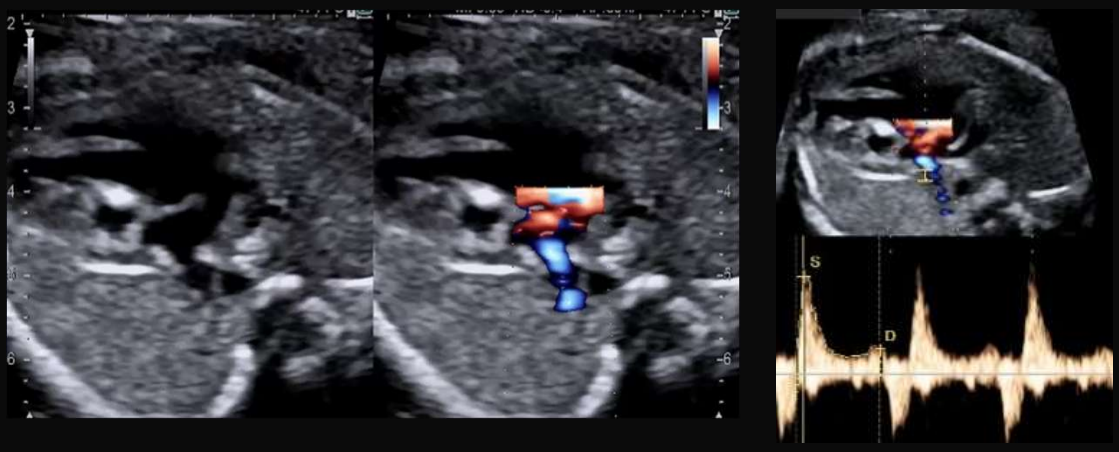
24W6D



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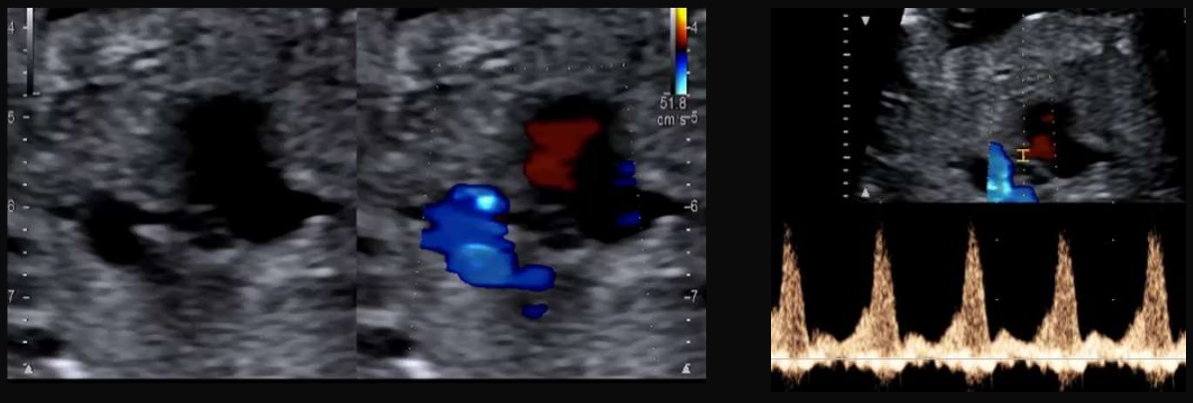
24W6D



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Key Points

- HLHS is a spectrum of complex cardiac malformations involving significant underdevelopment of the left ventricle and the left ventricular outflow tract.
- One of the most commonly diagnosed congenital heart abnormalities in utero
- LV is hypercontractile, small, or absent but can also be of normal size or dilated with no filling on color Doppler
- Aortic root is rudimentary and difficult to image on ultrasound
- Apex of the heart is predominantly formed by the right ventricle
- Foramen ovale bulges into the right atrium with left-to-right shunting on color Doppler

Abuhamad, A., & Chaoui, R. (2010). *A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts*. Lippincott Williams & Wilkins

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Key Points

- Pulmonary trunk and ductus arteriosus are compensatory dilated
- Reverse flow into the aortic arch in the 3VV
- Associated with a 4%-5% incidence of chromosomal abnormalities such as Turner syndrome, among others
- Extracardiac malformation have been reported in 10% - 25% of infants
- Presence of tricuspid valve dysfunction and/or restriction of flow across the foramen ovale cast a poor prognostic outlook
- Prenatal diagnosis has been associated with a lower incidence f perioperative neurologic events in some series.

Abuhamad, A., & Chaoui, R. (2010). *A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts*. Lippincott Williams & Wilkins