Comprehensive Echocardiography and Diagnosis of Major Common Congenital Heart Defects

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Supplementary Information The online version contains supplementary material available at https://doi.org/10.1007/978-3-031-26538-9_9.

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Y. Singh et al. (eds.), *Point-of-Care Ultrasound for the Neonatal and Pediatric Intensivist*,
https://doi.org/10.1007/978-3-031-26538-9_9

Introduction

Congenital heart diseases (CHD) are structural anomalies of the heart and great vessels. They represent the most frequent congenital anomalies with an incidence of 0.8–1 per 100 live newborns [1, 2]. Every year one million children worldwide are born with CHD and they are the first cause of mortality due to congenital anomalies [3, 4]. CHD develops early in fetal life as the heart is the first functional organ in the embryo. It is therefore of paramount importance to understand basic cardiac embryology and its disruption leading to CHD.

Basic cardiac embryology—a brief overview is summarized below and illustrated in Fig. 1 [5, 6].

(a) ***Formation of the heart tube***

In the second week of gestation, the human embryo consists of a disc within the amniotic fluid. Cardiogenic precursors in the epiblast will migrate through the primitive streak to form the mesodermal bilateral cardiogenic areas. These will merge cranially to form a horseshoe-shaped field. The embryonic disc will then undergo lateral folding, bringing together the two precursor areas creating the primitive heart tube. The heart starts to beat around day 21. From superior to inferior, the heart tube consists of the aortic sinuses, the truncus arteriosus, the bulbus cordis, the ventricle, the atrium, and the sinus venosus.

(b) ***Looping***

At day 23 the heart tube begins to loop with the bulbus cordis moving ventrally, caudally, and to the right (d-loop) and the primitive ventricle moving dorsally, cranially, and to the left.

(c) ***Septation***

Septation occurs between the fourth and fifth week of development. Two endocardial cushions develop from the dorsal and ventral surface of the atrioventricular canal and fuse, separating the atrium from the ventricle. Two other endocardial cushions on the lateral walls will ultimately form the tricuspid and mitral valve.

Septation of the atria begins with membranous tissue, the septum primum, growing from the roof of the atrium moving towards the endocardial cushions. Perforations in the center of the septum primum give rise to the foramen secundum. A muscular septum secundum grows to the right of the septum primum and will overlap the foramen secundum gradually. The remainder of the opening is called the foramen ovale.

The septation of the ventricles starts with a muscular interventricular ridge developing at the apex and ultimately fusing with the endocardial cushions.

(d) ***Systemic and pulmonary veins***

The right horn of the sinus venosus increases giving rise to the superior vena cava (SVC) and the inferior vena cava (IVC), the left sinus horn regresses and ultimately will become the coronary sinus. The primordial pulmonary vein is formed in the dorsal wall of the left atrium (LA) and the branches of the pulmonary veins become incorporated into the LA.

(e) ***Outflow tracts***

Neural crest mesenchymal cells in the bulbus cordis proliferate during the fifth week and form a bulbar ridge which continues in the truncus arteriosus. These cells migrate to reach the outflow tract. The ridges operate a 180-degree spiral movement to form the aortopulmonary septum which will then divide into the aorta and pulmonary trunk.

(f) ***Heart valves***

The atrioventricular valves develop between the fifth and eighth week of gestation. The left atrioventricular valve, the mitral valve has an anterior and posterior leaflet, the right atrioventricular valve, the tricuspid valve also has a septal leaflet. The valves are attached to the septum by thin fibrous chords inserted into the papillary muscles. The semilunar valves (aortic and pulmonary valves) are formed from the bulbar ridges and subendocardial tissue.

(g) ***Arterial system***

The arterial system consists initially of bilateral symmetric aortic arches which will undergo major changes to create the great arteries.

(h) ***Conduction system***

Cardiac development is a highly regulated process implicating complex molecular pathways at each step of development. A multitude of genes involved in this process has been described (Fig. 2) [7].

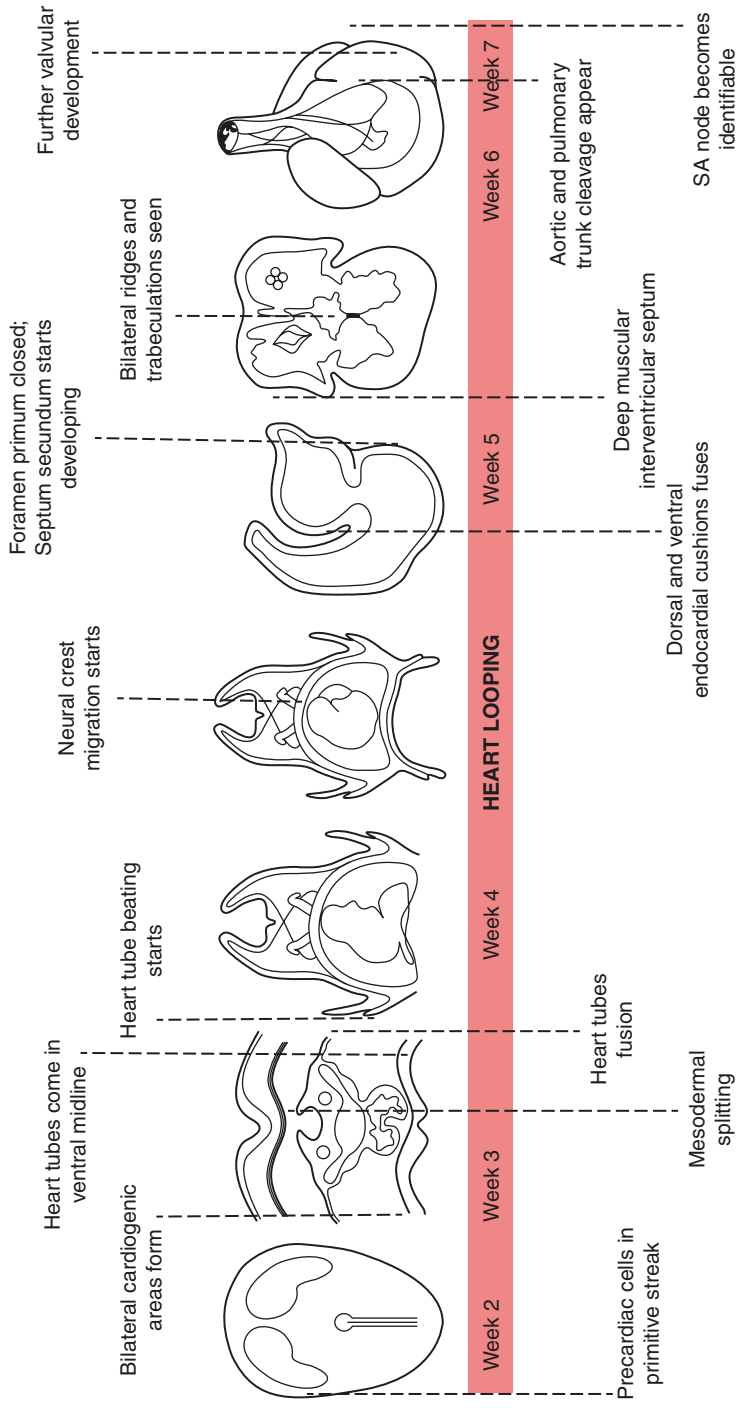

Fig. 1 Heart formation in the embryo (*Courtesy: Sheen Gahlaut and Yogen Singh*)

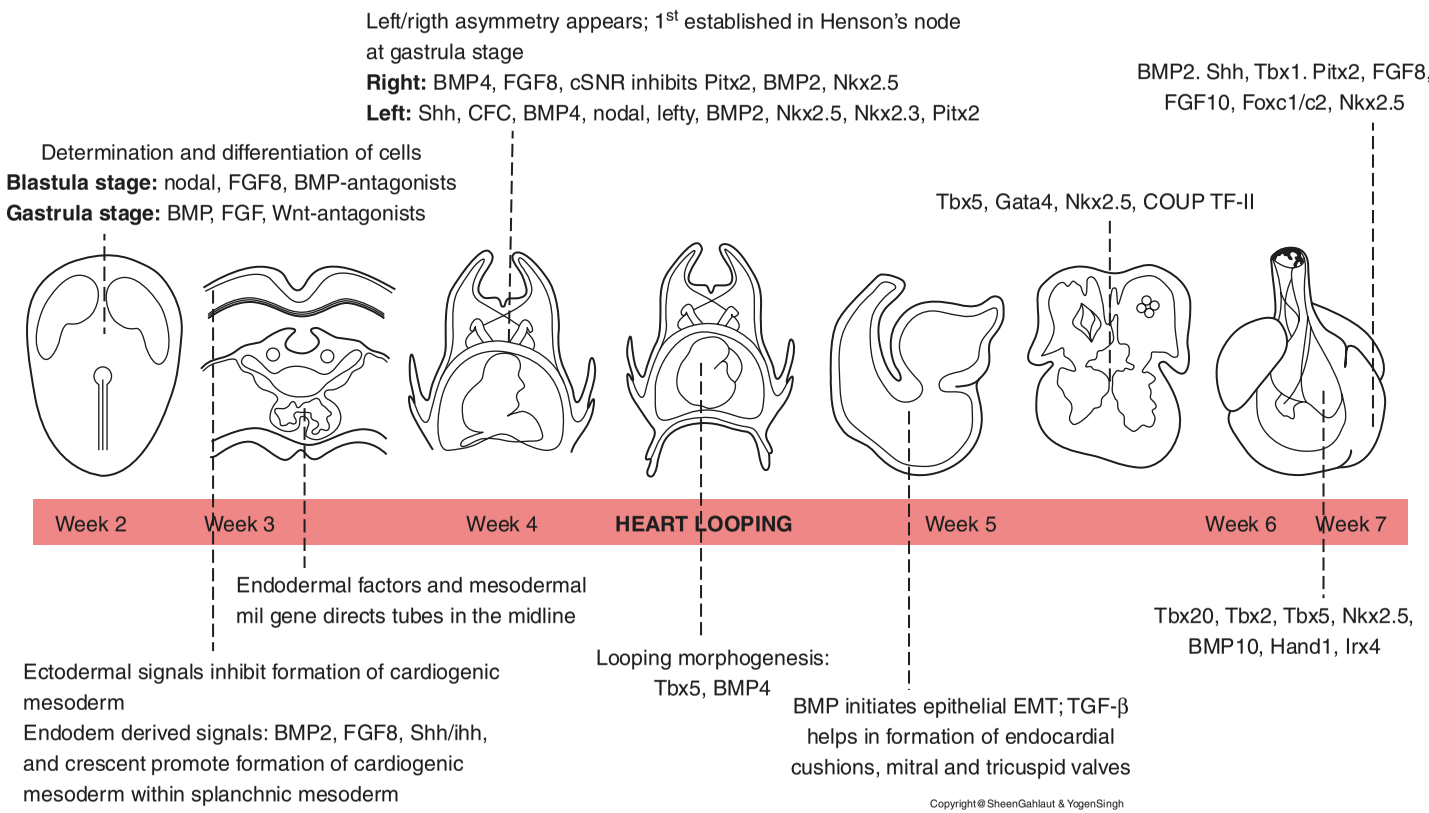

Fig. 2 Genes involved in cardiac development (*Courtesy: Sheen Gahlaut and Yogen Singh*)

Disruption of this process by different genetic, maternal, environmental, or most often unknown factors can lead to CHD in the fetus. During the heart developement, earlier the disruption occurs, the more severe the heart malformation will be [5, 6].

Classification of Congenital Heart Defects

To this date, there is no universally accepted classification for congenital heart defects, but the one most used is based on pathophysiology and it includes two major categories, non-cyanotic and cyanotic congenital heart defects (CHD). The non-cyanotic category can be subdivided into two groups: CHD with increased pulmonary blood flow and CHD with obstructive blood flow from the ventricles. The cyanotic category can be subdivided into CHD with decreased pulmonary blood flow and CHD with mixed blood flow [8, 9] (Fig. 3).

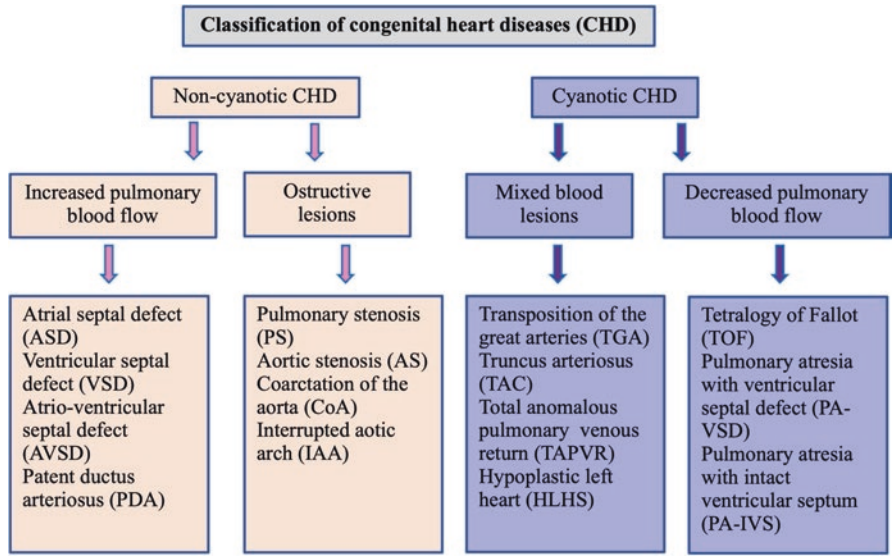

Fig. 3 A simple classification of congenital heart disease based on cyanosis and amount of pulmonary blood flow

Identifying Newborns with Critical Congenital Heart Disease

As many as 25–33% of infants born with CHD are considered to have critical CHD, which is defined as having a cardiac lesion requiring surgical or catheter-based intervention [3]. In these infants, any delay in diagnosis will increase morbidity and mortality [10, 11]. It is therefore important for neonatologist or intensivist performing echocardiography to be able to recognize these defects as early as possible.

Neonatologist performed echocardiography (NPE) or targeted neonatal echocardiography (TNE) should be performed by accredited neonatologists according to the guidelines of the European Society of Pediatric and Neonatal Intensive Care (ESPNIC) and American Society of Echocardiography (ASE)/Association for European Pediatric Cardiology (AEPC) [12, 13] (Tables 1, 2, and 3).

Table 1 Indications for performing NPE include (but not exhaustive list as there may be many more indications depending upon the clinical indications and NPE service availability):

Suspicion of PDA in premature baby (24–72 h of life)
Evaluation of a perinatal asphyxia
Abnormal cardiovascular adaptation in the first 24 h of life
Suspicion of persistent pulmonary hypertension
Congenital diaphragmatic hernia
Shock

Table 2 The first echocardiography should include a complete morphologic and functional examination using a segmental approach [14–17]

Atrial situs and position of the heart in the thorax
Systemic venous return
Size and morphology of the atria
Presence of atrial communication and direction of shunt
Atrioventricular connection and function of AV valves
Ventricular morphology, size, and function
Ventricular septal anatomy
Ventriculo-arterial connection and function of semilunar valves
Presence/absence of PDA and shunting
Coronary anatomy
Aortic arch and pulmonary artery anatomy
Pulmonary venous return

Table 3 The standard echocardiography required for NPE core examination

1. Anatomy
(a) Cardiac anatomy including
• Inflow
• Outflow
• Cardiac valves
• Cardiac chambers
(b) Skills
• 2D images of the neonatal heart in long axis, short axis, high parasternal, PDA and aortic arch view, apical and subcostal
• Mode M to measure LA/Ao ratio
• Pulsed and color Doppler to demonstrate normal blood flow across valves and outflow tracts
• Continuous Doppler (CW) to measure tricuspid regurgitation (TR)
2. Systolic LV function
(a) End-diastolic and end-systolic dimension of the LV (2D or M-Mode)
(b) End-diastolic and end-systolic thickness of the posterior LV wall (2D, M-Mode)
(c) End-diastolic and end-systolic thickness of the interventricular septum (2D, M-Mode)
(d) Shortening fraction (M-Mode)
(e) Ejection fraction (M-Mode or 2D Simpson)
3. Diastolic LV function
(a) Mitral valve max velocity of E wave (PW Doppler)
(b) Mitral valve max velocity of A wave (PW Doppler)
4. Evaluation of pulmonary hypertension
(a) Max velocity of TR (CW Doppler)
(b) End-diastolic velocity of pulmonary regurgitation (PR) (PW/CW Doppler)
5. Evaluation of PDA
(a) Minimal dimension of the PDA (2D)
(b) Shunt direction (color Doppler, PW, CW)
(c) Max and mean gradient of ductal flow (CW, PW)
6. Evaluation of an interatrial shunt:
(a) Direction of shunt (color Doppler)
7. Evaluation of pericardial effusion
(a) Measure of the effusion in diastole (2D)

Compared to comprehensive NPE evaluation, cardiac POCUS assessment is limited and focused at answering specific clinical question or target specific intervention. Indications for the cardiac POCUS, especially in neonates, are limited and it should NOT be used as a screening tool for the CHDs, although abnormality can be detected while performing cardiac POCUS for

other indications. If any CHD or cardiac abnormality suspected on cardiac POCUS performed by the neonatologist or intensivist, these cases should be urgently discussed with the pediatric cardiology service for a formal structural echocardiography and cardiac consultation. Although cardiac POCUS is not aimed at screening or diagnosing CHDs, still its important for the neonatal and pediatric intensivist performing cardiac POCUS to have a good knowledge of cardiovascular physiology and echocardiographic aspects of critical CHDs.

The majority of newborns with critical CHDs present with the one of the following 3 clinical presentations: 1) shock, 2) cyanosis, and 3) tachypnea (or respiratory symptoms). Each presentation is associated with certain types of CHDs. Infants with critical CHDs can be asymptomatic or can present with non-specific signs and symptoms, especially early in the clinical course while ductus arteriosus is still patent and / or pulmonary vascular resistance is high. Specific CHD will have some key echocardiographic features helping to pinpoint the diagnosis which will then have to be precisely determined on a comprehensive echocardiography by the pediatric cardiologist.

A summary of the most frequent critical CHD according to clinical symptoms, with their echocardiography features and best echocardiography views to suspect the diagnosis have been described below.

1. Shock (The Grey Neonate)

The main clinical signs will be poor peripheral perfusion, decreased or absent pulses, tachycardia, tachypnea, and respiratory distress syndrome as the ductus arteriosus closes and systemic perfusion decreases [18].

Main Cardiac Lesions

- Hypoplastic left heart syndrome
- Critical aortic stenosis
- Coarctation of the aorta
- Interrupted aortic arch

Common Key Echocardiography Feature: Poorly Functioning Left Ventricle

HLHS Echo Features and Best Views (Figs. 4, 5, 6, and 7; Videos 1 and 2)

Key echo feature: hypoplastic left ventricle

	PLAX	PSAX	A4/5C	SC	SS
Very small hyperechogenic ventricle with poor contraction	X	X	X	X	
No or very reduced flow through mitral valve	X		X		
No or very minimal flow through aortic valve	X		X	X	X
Small LA	X		X		
Small aortic annulus, very small ascending aorta, and arch	X		X		X
Retrograde flow in ascending aorta					X
PDA with right to left flow					X
Left to right shunt through PFO, sometimes restrictive (high velocity)				X	

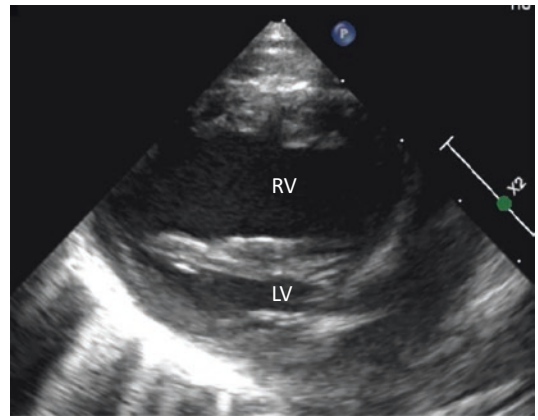

Fig. 4 PLAX: hypoplasia of the LV, dilated RV

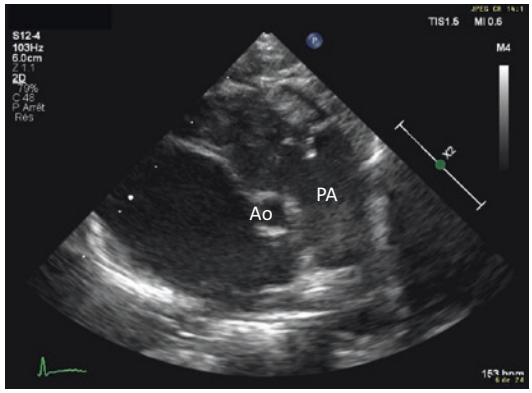

Fig. 5 PSAX : very small aorta (Ao), dilated pulmonary artery (PA)

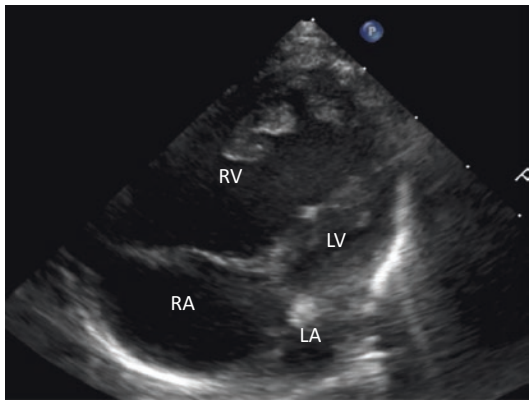

Fig. 6 4C: dilated RA and RV, hypoplastic LV, small LA, mitral atresia

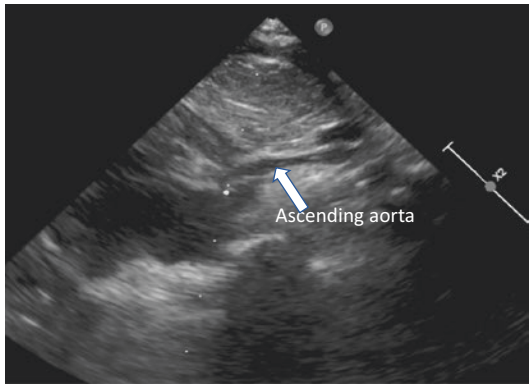

Fig. 7 Suprasternal: very hypoplastic ascending aorta

Critical Aortic Stenosis (Figs. 8 and 9; Videos 1–4)

Key echo feature: minimal antegrade flow through aortic valve

	PLAX	PSAX	A4/5C	SC	SS
Small or normal sized aortic annulus	X				
Very thickened aortic leaflets with decreased mobility	X	X			
Dilated, poorly contractile LV	X	X	X	X	
Hyperechogenic endocardium (endocardial fibroelastosis)	X	X	X	X	
Mitral regurgitation, dilated LA	X		X		
In severe cases retrograde flow in ascending aorta					X
Pulmonary hypertension	X		X		
Accelerated L-R shunt through PFO					X

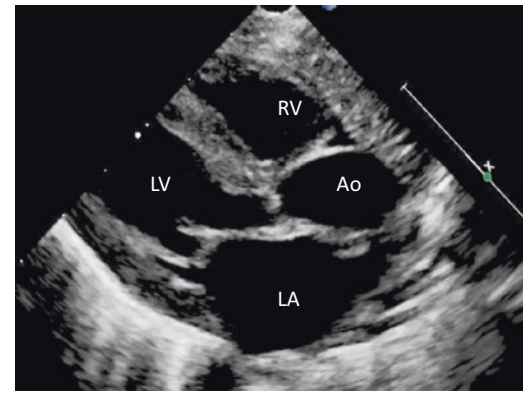

Fig. 8 PLAX: thickened aortic valve, post-stenotic dilatation of the ascending aorta, globular dilated LV with hypertrophy

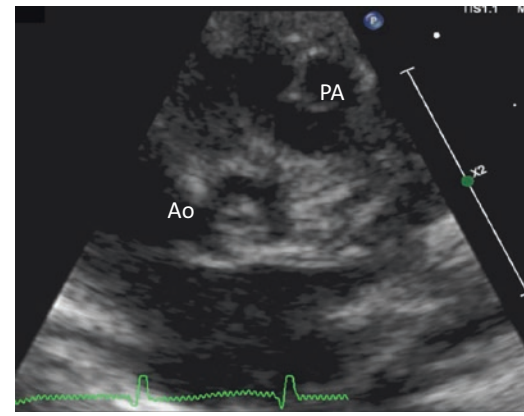

Fig. 9 PSAX: thickened aortic valve with minimal opening

Critical Aortic Coarctation (Figs. 10 and 11; Video 5)

Key echo feature: accelerated flow in descending aorta with run-off in diastole

	PLAX	PSAX	A4/5C	SC	SS
Dilated, poorly contractile LV	X	X	X	X	
Hypoplastic transverse aortic arch					X
Hypoplastic aortic isthmus					X
Bicuspid aortic valve		X			
PDA with left to right shunt		X			
Pulmonary hypertension	X	X	X		

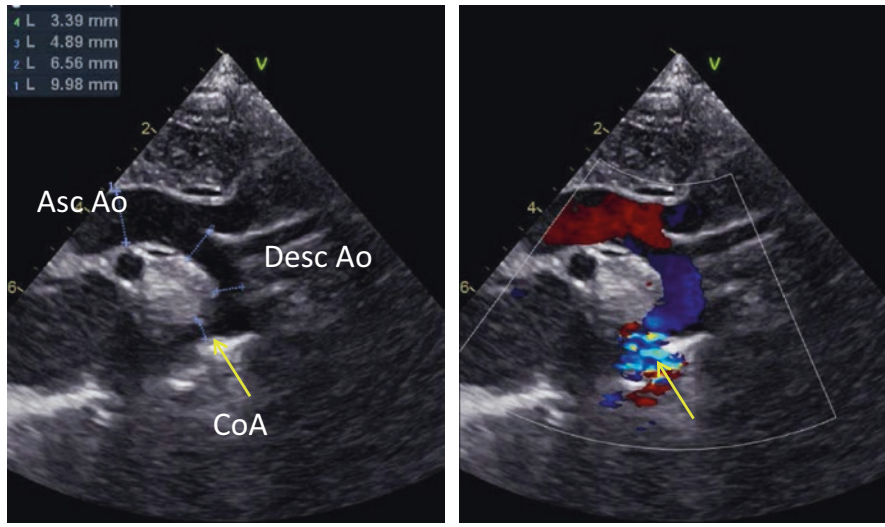

Fig. 10 Suprasternal: narrowing of the descending aorta (arrow = coarctation) in juxtaductal position with aliasing of flow by color Doppler

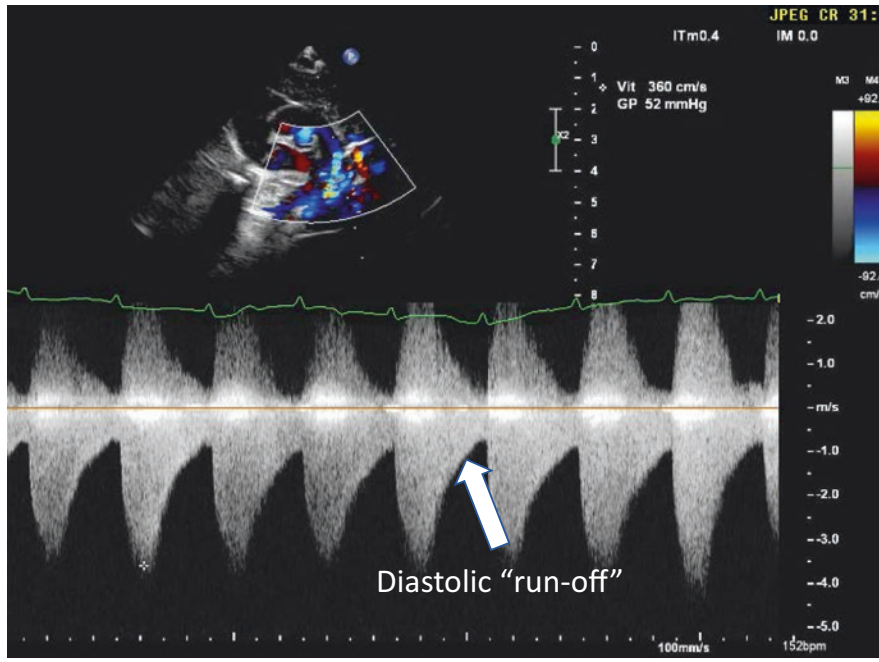

Fig. 11 Suprasternal Doppler: high-velocity flow with diastolic "run-off"

Interrupted Aortic Arch (Fig. 12)

Key echo feature: unable to visualize entire aortic arch

	PLAX	PSAX	A4/5C	SC	SS
Dilated brachiocephalic artery					X
Inability to image the entire arch in suprasternal view					X
PDA with right to left shunt		X			X
VSD	X		X	X	

2. Cyanosis (The Blue Neonate)

The main clinical signs will be central cyanosis, sometimes associated with signs of shock.

Ductal and non-ductal dependent lesions can cause cyanosis in the newborn. In some cases, there will be differential cyanosis with lower saturations in the lower extremities compared to the upper extremities (left heart obstructive lesions) or reverse differential saturation with higher saturations in the upper extremities compared to the lower extremities (Transposition of the great vessels with coarctation and pulmonary hypertension) [19, 20].

Ductal-Dependent Lesions

- Right heart obstructive lesions: severe pulmonary valve stenosis or pulmonary atresia with intact ventricular septum (PA-VSD), tetralogy of Fallot's

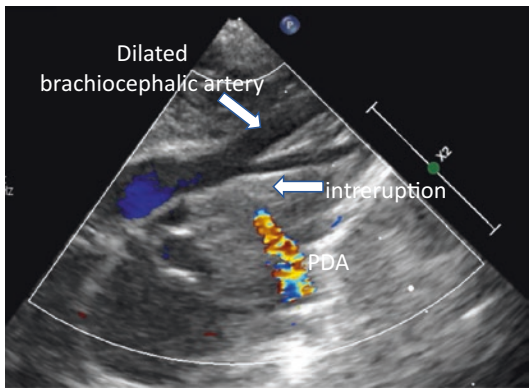

Fig. 12 Suprasternal: dilated brachiocephalic artery, descending aorta perfused through PDA

- Parallel circulation: transposition of the great arteries (TGA)

Severe Pulmonary Valve Stenosis (Fig. 13; Video 6)

Key echo feature: thickened and doming pulmonary valve with post-stenotic dilatation of pulmonary trunk

	PLAX	PSAX	A4/5C	SC	SS
Pulmonary valve thickened and doming, restricted opening		X		X	
Aliasing of flow into pulmonary artery		X		X	
Post-stenotic dilatation of pulmonary trunk		X		X	

Pulmonary Atresia with Intact Septum (Figs. 14, 15, and 16)

Key echo feature: hypoplastic, poorly contractile RV

	PLAX	PSAX	A4/5C	SC	SS
Small, hypertrophied RV, often not tripartite with decreased function	X		X	X	
Pulmonary valve thickened, restricted, or no opening		X		X	
No antegrade flow or very little antegrade flow into pulmonary artery		X		X	
Retrograde flow into pulmonary artery from PDA		X			X
Severe tricuspid regurgitation	X		X		
PFO with right to left shunt				X	

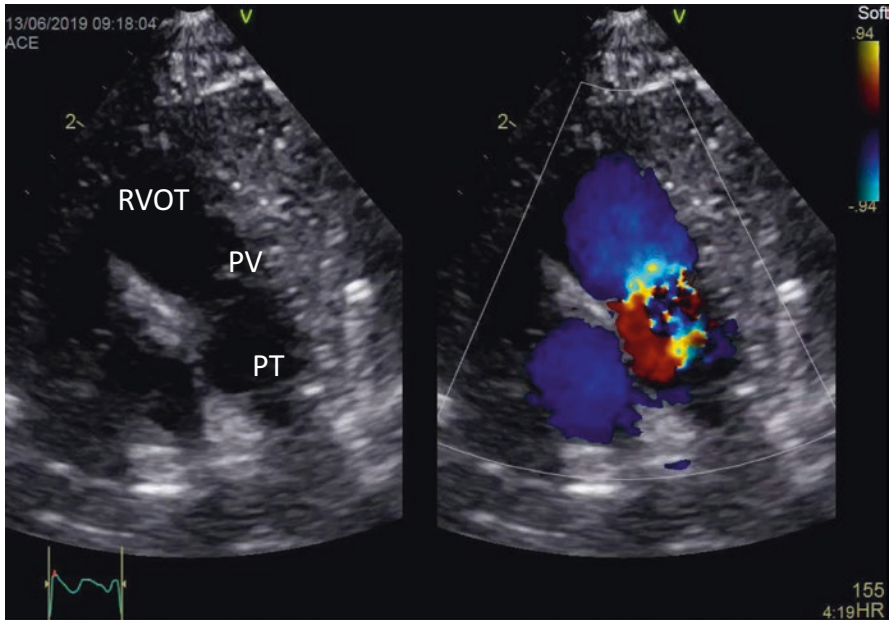

Fig. 13 PSAX: pulmonary valve (PV) thickened, doming with restricted opening and aliasing of flow, post-stenotic dilatation of pulmonary trunk (PT)

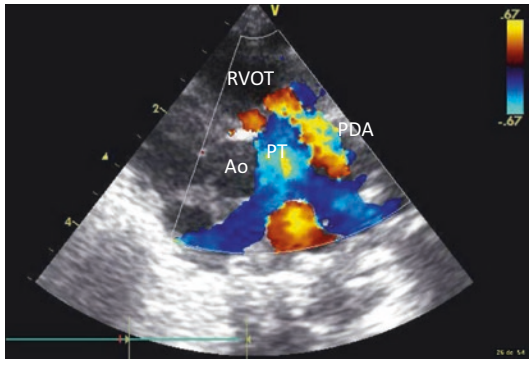

Fig. 14 PSAX: absent flow across pulmonary valve, retrograde flow in pulmonary trunk (PT) through patent ductus arteriosus (PDA)

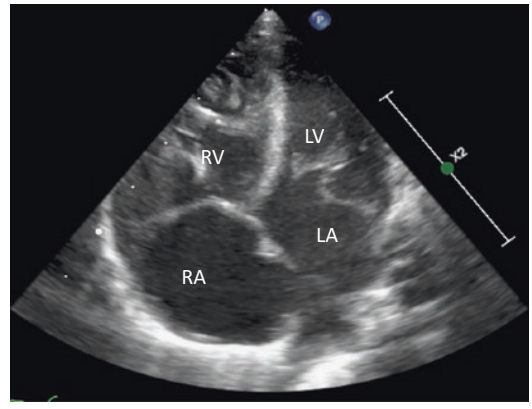

Fig. 15 4C: Hypertrophied and small RV, interatrial septum bulging to the left (and obligatory right to left interatrial shunt)

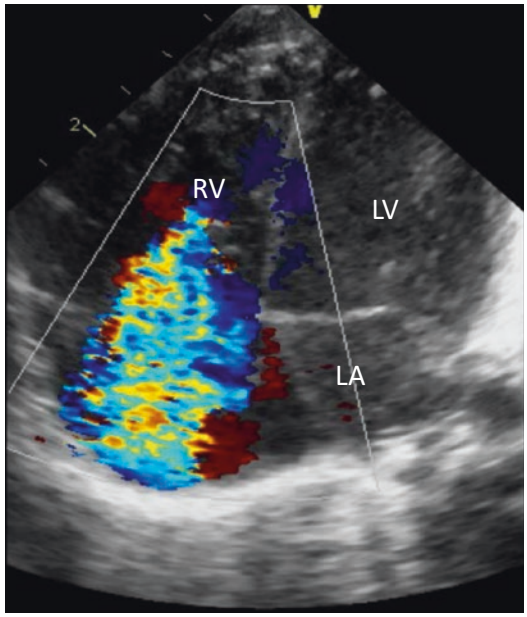

Fig. 16 4C: severe tricuspid regurgitation (TR)

Transposition of the great arteries (Figs. 17, 18, and 19)

Key echo feature: parallel arrangement of great vessels

	PLAX	PSAX	A4/5C	SC	SS
RV giving rise to straight vessel (aorta)	X			X	
LV giving rise to vessel bifurcating (pulmonary artery)	X			X	
Parallel arrangement of great vessels (cannonball)	X			X	
Aorta anterior and to left of pulmonary artery		X			
PDA with left to right flow		X			X

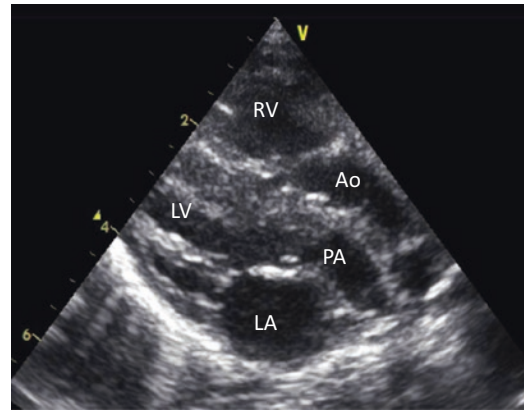

Fig. 17 PLAX: Pulmonary artery (PA) coming from LV, aorta (AO) coming from RV, parallel arrangement of the great vessels

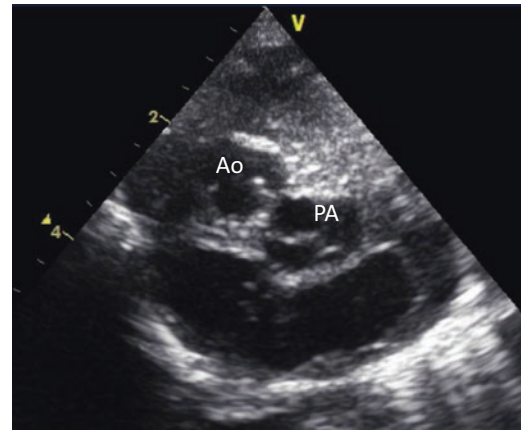

Fig. 18 PSAX: Aorta anterior right, pulmonary artery (PA) posterior left

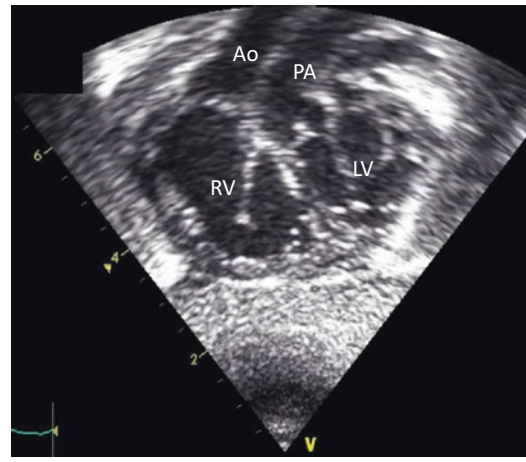

Fig. 19 Subcostal: Parallel arrangement of the great vessels, aorta anterior right and pulmonary artery (PA) posterior left bifurcating into branches

	PLAX	PSAX	A4/5C	SC	SS
PFO with bidirectional flow				X	

Non-ductal-Dependent Lesions

- Truncus arteriosus
- Tetralogy of Fallot
- Total anomalous pulmonary venous return
- Tricuspid atresia

Truncus Arteriosus (Figs. 20 and 21; Video 7)

Key echo feature: only one great vessel exiting heart

	PLAX	PSAX	A4/5C	SC	SS
Large perimembranous VSD	X		X	X	
Single large arterial vessel overriding VSD giving rise to aorta and pulmonary arteries	X	X		X	
Absent PDA		X			X
Truncal valve thickened with stenosis and/or regurgitation, sometimes quadricuspid		X			

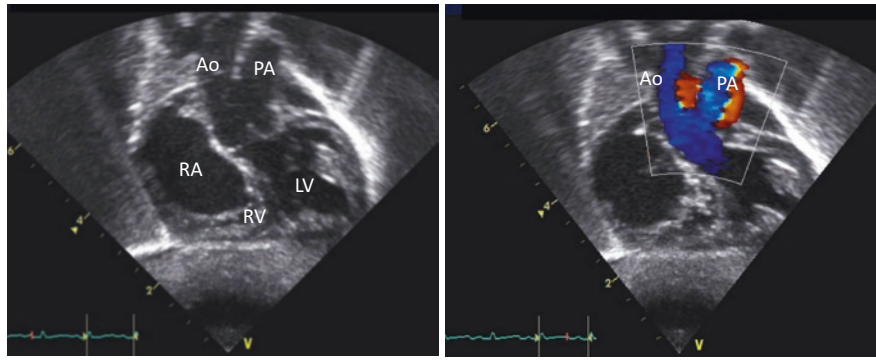

Fig. 20 Subcostal: Single artery coming from LV giving rise to aorta and pulmonary artery (PA)

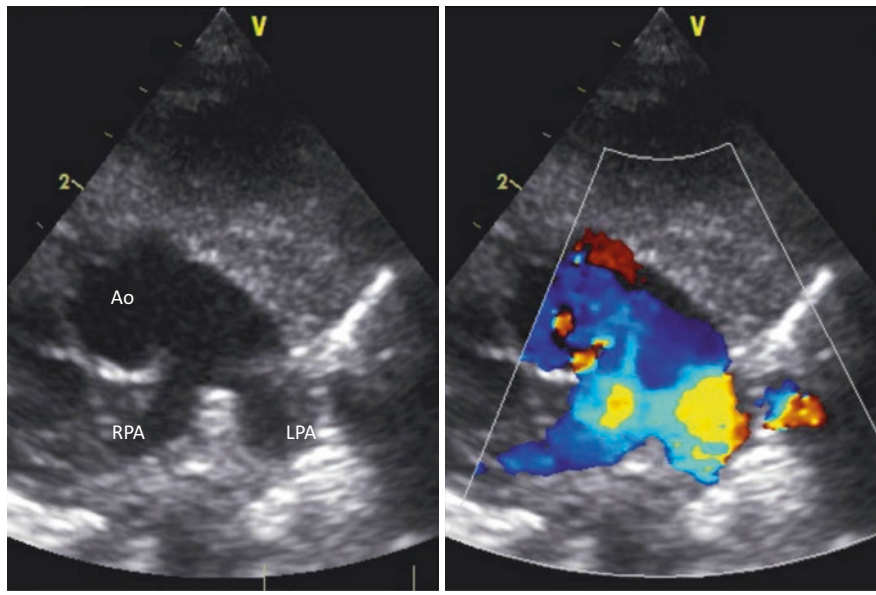

Fig. 21 PSAX: Branch right (RPA) and left (LPA) pulmonary arteries coming from aorta (AO)

Tetralogy of Fallot (Figs. 22 and 23; Video 8)

Key echo feature: VSD and overriding aorta

	PLAX	PSAX	A4/5C	SC	SS
Large perimembranous VSD	X		X	X	
Large overriding aorta	X	X		X	
RV hypertrophy		X		X	
Infundibular and valvar/supravalvar pulmonary stenosis		X		X	
Some degree of pulmonary hypoplasia		X		X	
Right aortic arch (25%)					X

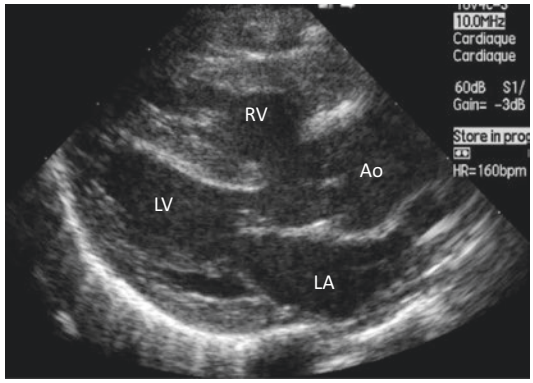

Fig. 22 PLAX: Large ventricular septal defect (VSD) and overriding aorta

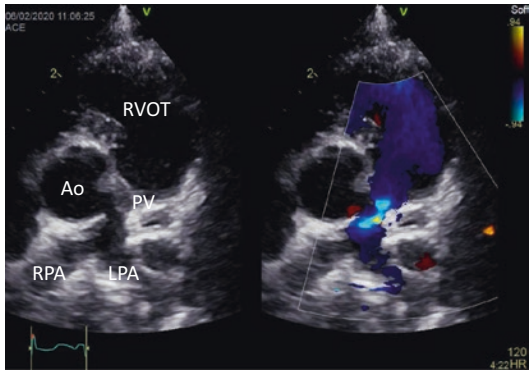

Fig. 23 PSAX: Hypoplasia of pulmonary valve (PV) annulus, trunk (PT), and branch left (LPA) and right (RPA) pulmonary arteries with aliasing of flow through PV

Total Anomalous Pulmonary Venous Return (TAPVR) (Figs. 24 and 25; Video 9)

Key echo feature: very small LA, right to left shunt through PFO

	PLAX	PSAX	A4/5C	SC	SS
Inability to visualize pulmonary veins entering LA			X		X
Very small LA	X		X	X	
Very dilated right-sided cavities	X	X	X	X	
PFO with right to left shunt				X	
Pulmonary hypertension		X	X		
Either dilated SVC, IVC, or coronary sinus			X	X	X
Vertical vein to innominate vein					X
Dilated portal vein (infradiaphragmatic TAPVR)				X	

Tricuspid Atresia (Fig. 26; Videos 10 and 11)

Key echo feature: only one AV valve present (left)

	PLAX	PSAX	A4/5C	SC	SS
Absent tricuspid valve (echogenic band)			X	X	
No flow through tricuspid valve			X		
Some degree of RV hypoplasia	X		X	X	
PFO with right to left shunt				X	
Associated with VSD, pulmonary stenosis/atresia, transposed great vessels depending on subtype	X	X	X	X	

3. The Tachypneic Neonate (Respiratory Symptoms)

Tachypnea is usually due to pulmonary edema secondary to increased pulmonary blood flow as pulmonary vascular resistance decreases after birth. The main clinical signs of increased pulmo-

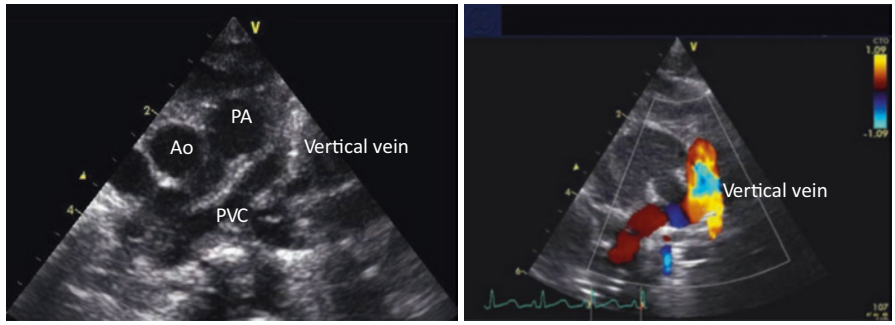

Fig. 24 High left parasternal: Pulmonary vein collector (PVC) and vertical vein (red flow, right-sided picture)

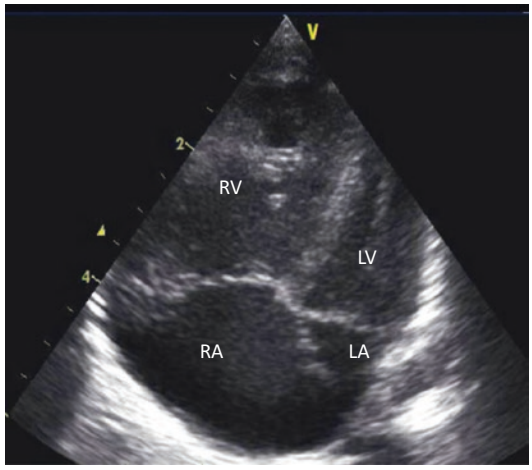

Fig. 25 4C: small LA, dilated RA and RV

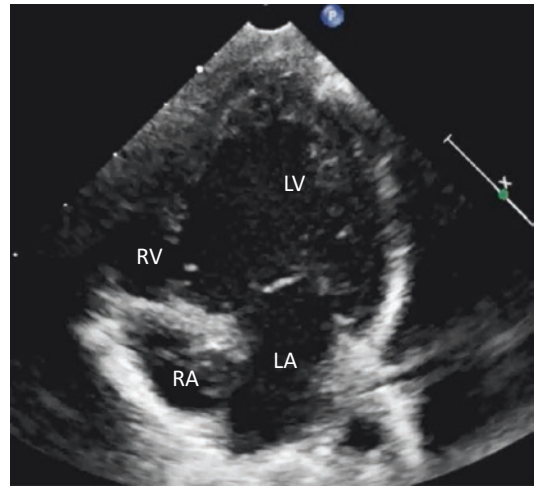

Fig. 26 4C: Echogenic band at place of tricuspid valve, hypoplastic RV

nary blood flow are tachypnea, increased work of breathing or respiratory distress. Respiratory distress can also be due to elevated pulmonary venous pressures or pulmonary venous congestion [21].

Main Cardiac Lesions

- Truncus arteriosus (cf above)
- Patent ductus arteriosus in premature infants
- Large ventricular septal defects
- Atrio-ventricular septal defects (AVSD)
- Total anomalous pulmonary venous return with obstruction - pulmonary blood flow is not increased in this lesion but obstruction leads to deranged pulmonary venous return and pulmonary venous congestion

Patent Ductus Arteriosus (Figs. 27 and 28; Video 12)

	PLAX	PSAX	A4/5C	SC	SS
Flow from descending aorta to pulmonary artery		X			X
Dilated LA/LV	X		X		
Retrograde flow in descending aorta					X
Retrograde flow in abdominal aorta				X	

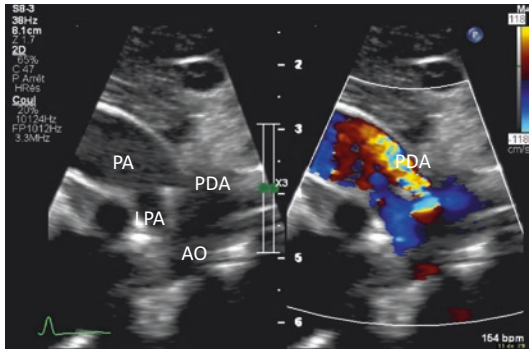

Fig. 27 PSAX: PDA is seen connecting the PA and the descending aorta

Ventricular Septal Defect (Figs. 29, 30, 31, and 32; Videos 13 and 14)

	PLAX	PSAX	A4/5C	SC	SS
Echolucent space in interventricular septum	X	X	X	X	
Left to right systolic flow through VSD	X	X	X	X	
Dilated LA/LV	X		X		

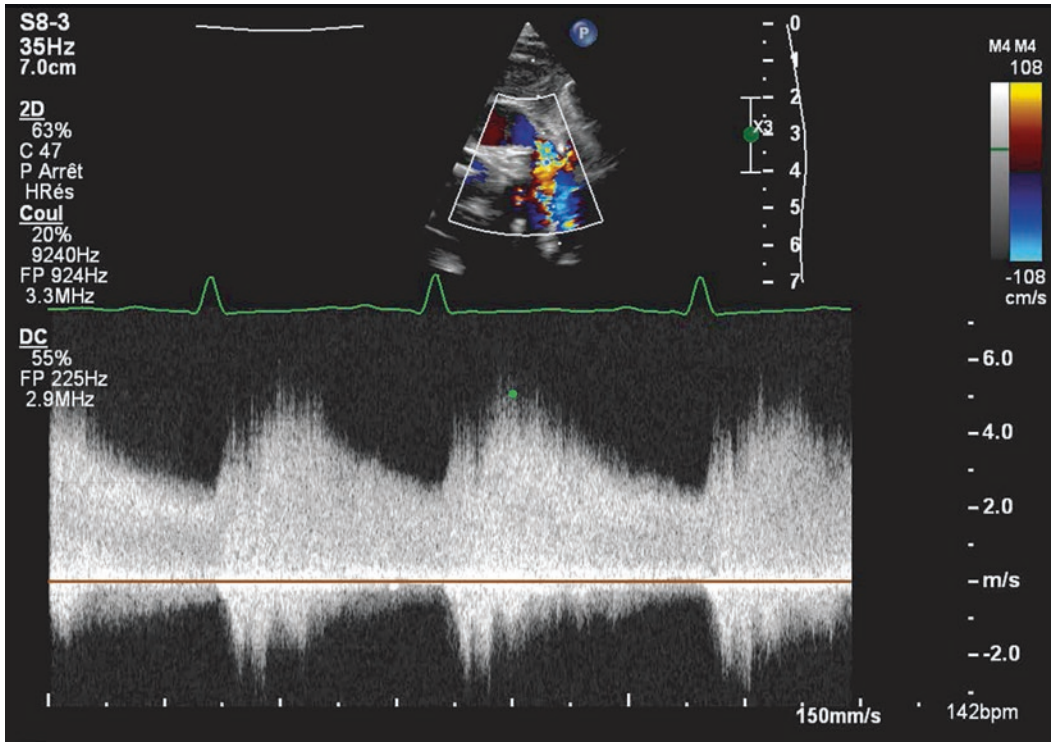

Fig. 28 Doppler high velocity left to right shunt (5 m/s) through the PDA, allowing to estimate a systolic pressure gradient of 100 mmHg (Bernoulli = $4 \times V^2$) between aorta and PA (restrictive PDA)

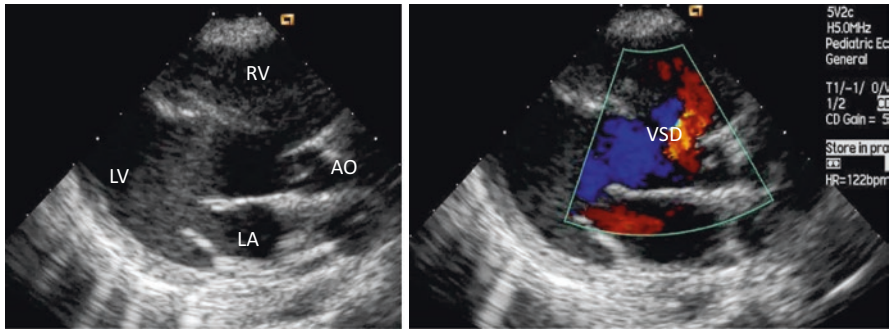

Fig. 29 PLAX: Echolucent space between the two ventricles = perimembranous VSD with low velocity left to right shunt

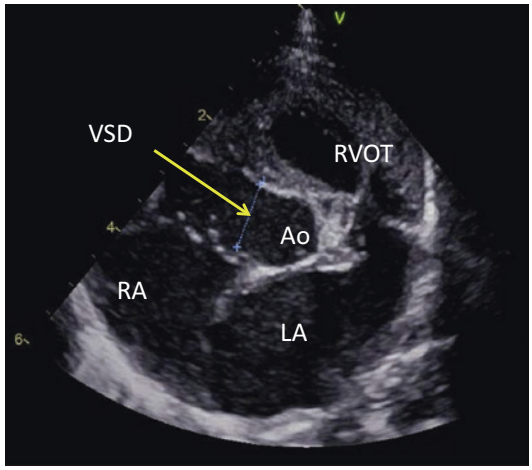

Fig. 30 PSAX: Echolucent space in the sub-aortic interventricular septum

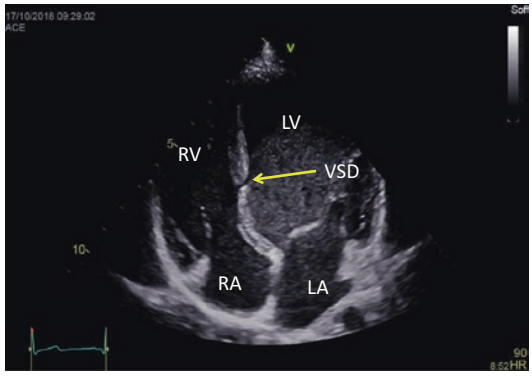

Fig. 31 4C: echolucent space in the muscular interventricular septum (arrow) = trabecular VSD with dilated LV

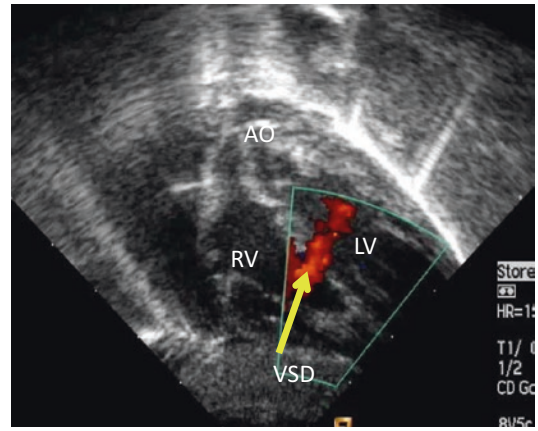

Fig. 32 Subcostal: left to right flow through muscular VSD

Atrial Septal Defect (Figs. 33 and 34; Videos 15 and 16)

	PLAX	PSAX	A4/5C	SC	SS
Echolucent space in secundum interatrial septum		X	X	X	
Left to right flow through VSD and ASD		X	X	X	
Aliasing of flow through pulmonary valve (PV)		X		X	
Dilated RV and RA	X	X	X	X	

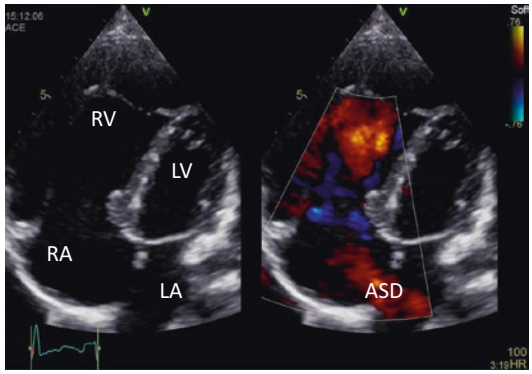

Fig. 33 4C: echolucent space in the interatrial septum with left to right shunt, dilated RA and RV

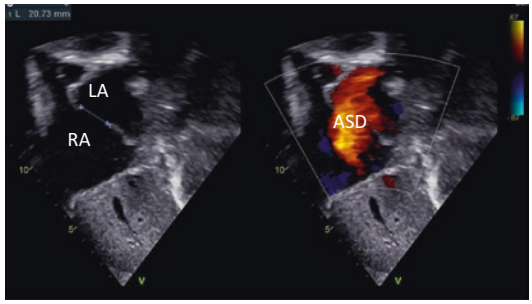

Fig. 34 Subcostal: left to right shunt through atrial septal defect (ASD)

Atrioventricular Septal Defect (Fig. 35; Video 17)

Key echo feature: crux of the heart defect

	PLAX	PSAX	A4/5C	SC	SS
Echolucent space in inlet interventricular septum		X	X	X	
Common atrioventricular valve	X	X	X	X	
Echolucent space in primum interatrial septum		X	X	X	
Left to right flow through VSD and ASD		X	X	X	
Dilated left and right heart chambers	X	X	X	X	
Atrioventricular valve regurgitation frequent	X	X	X	X	

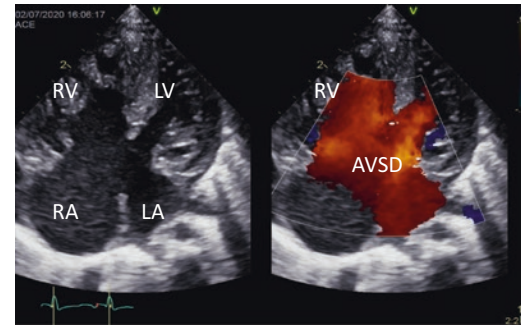

Fig. 35 4C: Echolucent space in primum atrial septum and inlet ventricular septum (AVSD: defect of the crux of the heart) with common atrioventricular valve

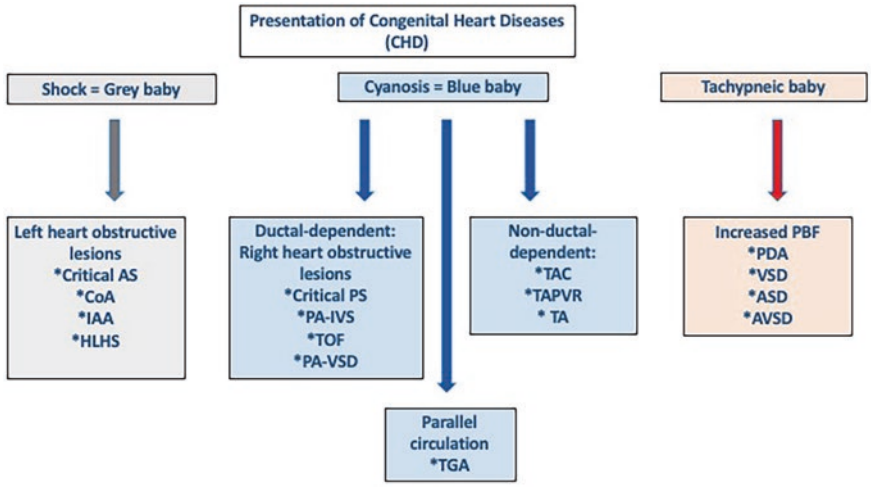

Fig. 36 Clinical presentation of congenital Heart Diseases (CHD), some CHD may show overlapping between the different categories. Abbreviations: *AS*: aortic stenosis; *ASD*: atrial septal defect; *AVSD*: atrio-ventricular septal defect; *CoA*: coarctation of the aorta; *HLHS*: hypoplastic left heart syndrome; *IAA*: interrupted aortic arch *PA-IVS*: pulmonary atresia intact ventricular septum; *PA-VSD*: pulmonary atresia with ventricular septal defect; *PBF*: pulmonar blood flow; *PDA*: patent ductus arteriosus; *PS*: pulmonry stenosis; *TA*: tricuspid atresia *TAC*: truncus arteriosus (common arterial trunk); *TAPVR*: total anomalous pulmonary venous return; *TGA*: transposition of the great arteries; *TOP*: Tetralogy of Fallot; *VSD*: ventricular septal defect

Conclusion

In well-trained hands, echocardiography is a great tool allowing the detection and diagnosis of almost all congenital heart defects. Early recognition of critical CHD may be lifesaving, especially for ductal-dependent lesions or for those when adequate mixing between the pulmonary and systemic circulation is crucial for survival. Some of these infants with inadequate central mixing of blood may need urgent atrial septostomy. A simple classification of critical CHDs based upon their clinical presentation is summarized in Fig. 36 above. When such lesions are encountered, prompt treatment should be initiated but for any newborn with suspected CHD, pediatric cardiology referral is mandatory in order to make the best medical and surgical therapeutic plan for the child.

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