Chapter

Fetal Echocardiogram Normal and Abnormal

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Abstract

In this chapter, the normal anatomy of the heart as well as pathologic cases is consistent with cardiac malposition and isomerism, septal defects, pulmonary stenosis/atresia/absent pulmonary valve syndrome, aortic malformation, hypoplastic left heart, conotruncal anomalies/common arterial trunk, tricuspid dysplasia, Ebstein anomaly, univentricular heart, and systemic venous abnormalities among other congenital cardio vascular defects by ultrasound images. Anatomical details of most CHD in fetus were provided by two-dimensional (2D) ultrasound with higher quality imaging, which enhances the diagnostic accuracy in a variety of CHD.

Keywords: congenital heart disease, fetal echocardiogram, ultrasound

1. Introduction

The incidence of congenital heart disease (CHD) is 8–9 per 1000 live births [1]. Including all subtle cardiac anomalies, the overall CHD incidence may be of 50 per 1000 live births (like bicuspid aortic valve, atrial septal aneurysm and Persistent left SVC) [2]. There are several risk factors for CHD which includes fetal and maternal factors. The fetal risk factors are extra cardiac abnormalities which are frequently associated with CHD even in the presence of normal karyotype [3]. About 10–20% of nonimmune hydrops is associated with CHD [4, 5]. One percent of arrhythmias are associated with CHD. Maternal factors like Diabetes mellitus, Phenylketonuria, Maternal exposure to Anticonvulsants, ethanol, nonsteroidal anti-inflammatory drugs, ACE inhibitors, indomethacin, and use of lithium increase the risk of CHD. IVF pregnancies increase CHD rates by four folds. CHD or syndromes associated with CHD in first-degree relatives increase risk of CHD. Half of the CHD cases are minor abnormalities and are easily corrected by surgery; prenatal diagnosis allows for better counseling and improved the outcome.

Guidelines for fetal echocardiogram by International society of ultrasound in obstetrics and gynecology (ISUOG), American institute of ultrasound in medicine (AIUM) and association for European pediatric cardiology (AEPC) (**Figure 1**): presence of clear guidelines for defining cardiac screening and fetal echocardiography helps to standardize the approach for the sonographic evaluation of fetal echocardiography.

Fetal echocardiogram is most optimally performed between 18 and 22 weeks gestation. The screening examination includes the upper abdomen; the four-chamber (4ch) view and outflow tracts for obtaining these planes need a sweep from four chamber view and into the upper mediastinum. Initial sweep shows five chamber (5ch) view followed by right ventricular outflow tract (RVOT), three vessel view

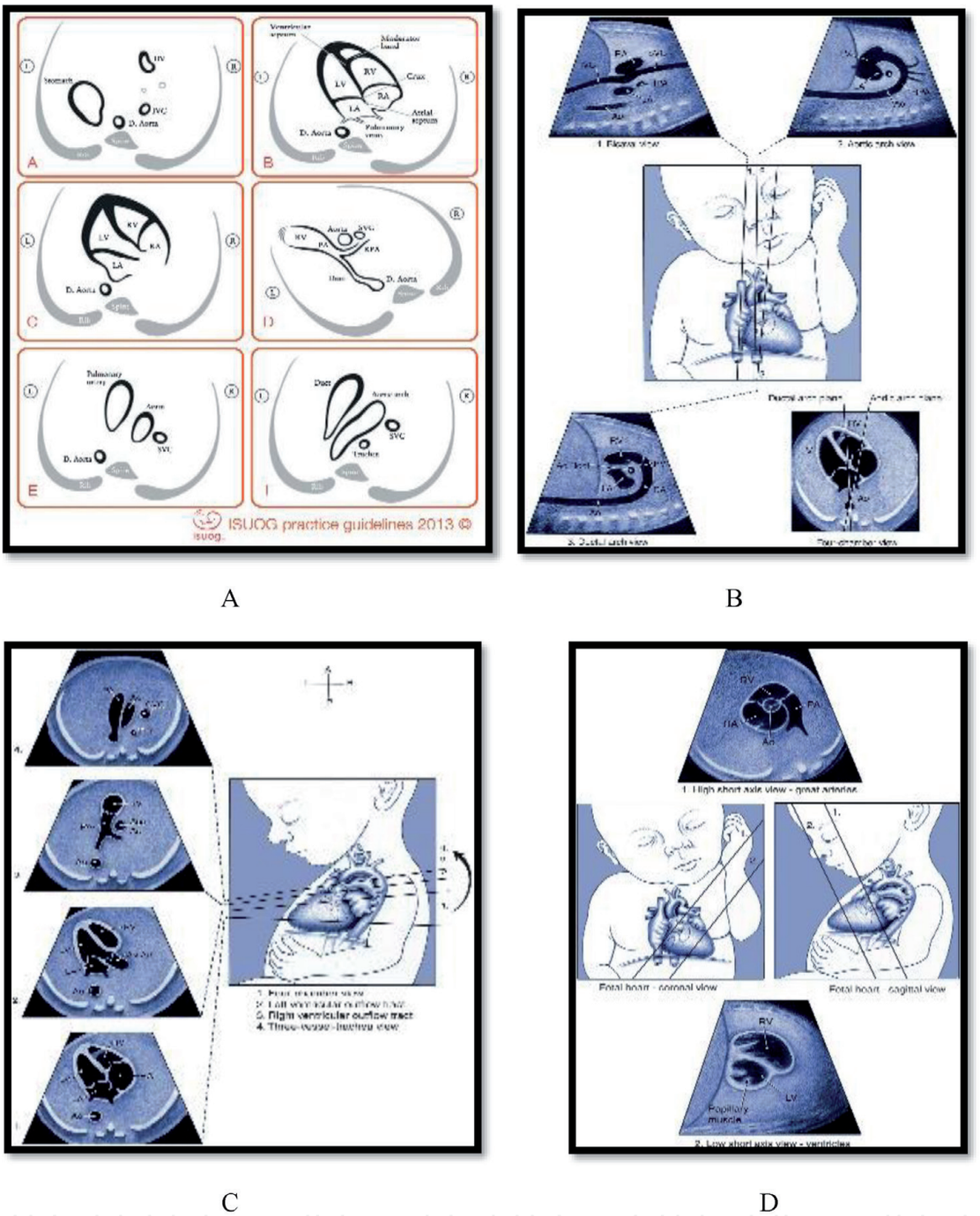


Figure 1.
(A) Selected views of fetal cardiac screening recommended by ISUOG. (B) Sagittal view as recommended by AIUM. (C) Standardized transverse scanning planes as recommended in the guidelines of the AIUM. (D) Low and high short axis views as recommended by AIUM.

(3vv) and three vessel trachea view (3VT) obese patients. Color flow Doppler may facilitate imaging of various cardiac structures and in obese patients. Guidelines involve situs determination and a sequential segmental analysis of Atria, Ventricles and great arteries and their connections. Documentation fetal heart rhythm should be performed. Fetal echo should be performed by real time imaging with acquisition of recording or digital video clips and still images. It is important to note that screening for cardiac malformations in the low risk population includes minimum views of four chambers and outflow tracts. It is not specified to screen three vessel trachea view, but it is relatively easy plane to obtain and provides significant information on outflow tracts and arches.

2. Embryology of heart

In third week of post conception, the embryo consists of three basic germ layers–the Ectoderm, Mesoderm and Endoderm. The lateral splanchnic mesoderm involves in the formation of single heart tube. The major steps in development of embryonic heart are (1) primitive heart tube, (2) looping, (3) atria, ventricles and outflow tracts septations, (4) septation of the atria, (5) septation of ventricles, (6) septation of the outflow tracts.

During cardiac looping, the primitive ventricles move downward to the right while the primitive atrium moves upward and to the left behind the ventricle. In the folded cardiac tube various regions are recognized and are separated by transitional zones. These regions will develop into cardiac cavities and the zones will become the septa and valves. Systemic veins begin with the formation of three paired veins the vitelline, umbilical and common cardinal veins, but the pulmonary veins develop separately.

3. Genetic diseases related to cardiac anomalies

CHD and chromosomal deletion syndrome: (1) DiGeorge syndrome or 22q11microdeletion; (2) Williams-Beuren syndrome, 1p36 deletion syndrome.

CHD associated with single gene disorders: Noonan syndrome and RASopathies, Holt-Oram syndrome, Alagille syndrome, Tuberous sclerosis complex and CHARGE syndrome.

Familial recurrence of congenital heart defects: recurrence risk of 3% for two healthy nonconsanguineous parents with one affected child, risk of recurrence increases to 10% with two affected siblings.

3.1 Fetal situs

Assessment of fetal visceral situs is essential first step in fetal echocardiogram. Evaluating cardiac position and orientation in thoracic cavity and anatomical relationship of abdominal organs is the part of the fetal echocardiogram examination.

Steps for sequential segmental analysis in the fetus are identifying visceral situs, Atrial arrangement, Atrioventricular valves, ventricular arrangement, ventriculoarterial connection, Ventricular outflow tracts and systemic and pulmonary venous connections. First step is Fetal visceral situs (**Figure 2**) for this identify fetal presenting part (cephalic/breech) next step is to determine fetal lie by obtaining sagittal view of fetal spine and compare with the maternal spine after this determine the location of the fetal left side with the regard to maternal abdomen. Bronshtein et al. [6] described a method to determine situs is referred as the right-hand rule for abdominal scan and left-hand rule for transvaginal approach. Palm of the hand corresponds to the face of fetus and sonographer holds the hand according to the side of fetal face; left side of the fetus will be shown by the examiners thumb.

Types of visceral situs are situs solitus, situs inversus and situs ambiguous.

Situs solitus is normal viscerocardiac arrangement where the cardiac apex, stomach and descending Aorta should be located on the fetal left side. Inferior vena cava is located on the right side. Situs inversus is the mirror image arrangement of organs and vessels to situs solitus. Incidence is about 0.01%. There is slight increased risk of CHD and 20% association with Kartagener's syndrome which is autosomal recessive transmission.

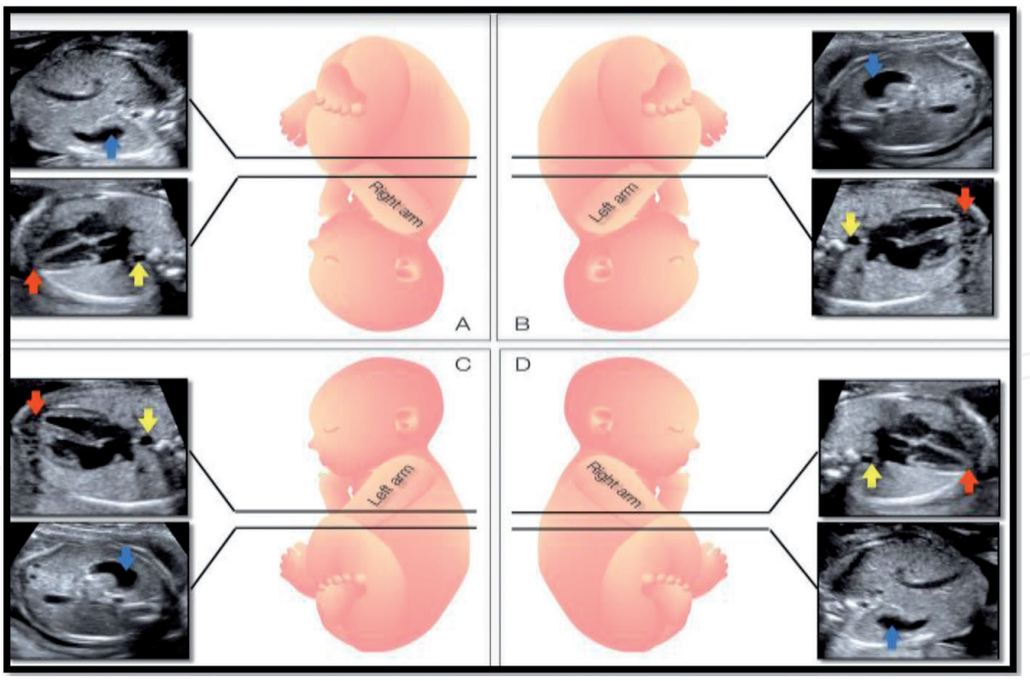


Figure 2.
Determining the fetal situs in longitudinal lie. (A) Fetus in cephalic presentation with spine close left uterine wall, results right side being anterior and left side posterior. (B) Cephalic with spine close to left uterine wall results left side being anterior and right side posterior. (C) Fetus in breech presentation with spine close to left uterine wall results left side being anterior and right side posterior. (D) Fetus in breech with spine close to right uterine wall results right side being anterior and left side posterior. The corresponding transverse ultrasound planes of the chest and abdomen. Blue arrow indicates fetal stomach, red arrow apex of heart and yellow arrow descending aorta.

Situs ambiguous, which refers to Viscerocardiac malposition, is commonly associated with complex CHD. Incidence of situs ambiguous is around 1per 10,000 infants [7]. This may be of right or left isomerism or situs ambiguous also known as visceral Heterotaxy syndrome which is very less commonly noted.

Detection of CHD on ultrasound depends on the position and axis of the cardia. Fetal cardiac axis: to determine the axis, obtain the transverse view of the chest at the level of four chamber view plane. A line is drawn from the spine to the anterior chest wall, the cardiac axis is the angle that the interventricular septum makes with this line (**Figure 3A**). Normal angle lies 45° to the left of the midline [8]. It is independent of gestational age. The axis is abnormal when it is >65° and < 25°. Study of Smith et al. [9] showed that >75° of leftward deviation are associated with CHD in 76% of fetuses which include Tetralogy of Fallot, Common arterial trunk, coarctation of aorta and Ebstein anomaly. Double outlet right ventricle, Atrioventricular septal defect and common atrium are more commonly associated with right axis deviation.

Cardiac position: depending upon the position of the heart it can be described as Levocardia Dextrocardia and mesocardia. Ectopia cordis refers when the cardia is outside the chest. As per Abuhamad et al [10] Heart positioned in right chest regardless of its axis it is termed as Dextrocardia, when the heart is placed in right chest with axis pointing to left then it is termed as Dextroposition and when the heart is positioned in right chest with axis pointing toward right side is known as dextroversion. Dextrocardia with axis to right: more commonly associated with Situs inversus, Congenital corrected transposition. Whereas Dextrocardia with left axis deviation is more commonly associated with extrinsic factors (**Figure 3B** and **C**) resulting in shift of heart to right side like Left diaphragmatic hernia, left lung mass, left pleural effusion, agenesis or hypoplasia of the right lung (scimitar syndrome). Mesocardia-Heart located in central chest

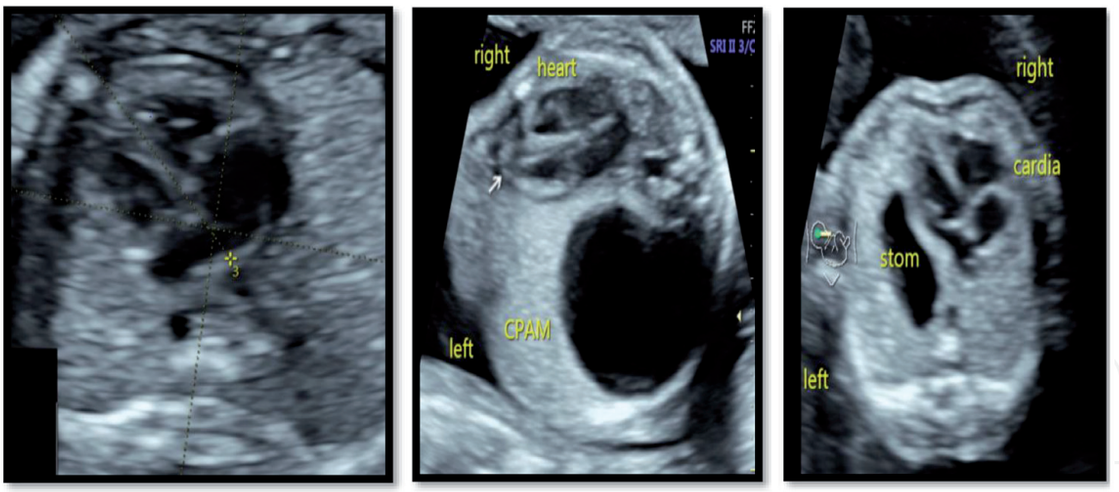


Figure 3.
(A) Measuring the cardiac axis. (B) Dextrocardia due to congenital adenomatoid malformation of left lung. (C) Dextrocardia with apex pointing to left due to congenital diaphragmatic hernia.

with cardiac apex pointing toward the midline of the chest usually associated with abnormal ventriculoarterial connections such as transposition of great arteries, Double outlet right ventricle. Bilateral increased lung volume such as laryngeal atresia is also associated with mesocardiac. Levocardia–it is more commonly used term for normal position of heart. Levoposition is noted in Right sided space occupying lesions or agenesis/hypoplasia of left lung. Levocardia with left axis deviation is commonly associated with CHD. Normal heart size should be 1/3rd to 1/2 that of thoracic cavity. The width of heart at AV valves corresponds to gestational age in weeks. Cardiothoracic (C/T) circumference is constant throughout gestation is about 0.45–0.5. Contractility of vetricles should be equal. Rhythm normal is 120–160 beats per minute.

4. Optimization of two-dimensional grayscale image in fetal echocardiography

The quality of 2D image is dependent on several factors such as choice of the transducer and presets, the angle of insonation and access to the region of interest and magnification of target region. Routinely two types of transducers are used in fetal echocardiography low frequency range (2–5 MHz) which allows good penetration and acceptable resolution. High frequency range (5–8 MHz) allows for improved resolution but limited penetration. Recently linear transducers are used for first trimester echocardiography as it gives high resolution. Image presets: (1) Harmonic imaging: in harmonic imaging, the reflected harmonic wave has a low amplitude high frequency which results in improved image and contrast with reduced artifacts. (2) Compound imaging allows the transducer to send signals at multiple angles to eliminate artifacts and improves the image resolution. (3) Speckle reduction imaging: weak signals are eliminated and strong signals are enhanced this allows the image to become smoother and reduces artifacts. (4) Focal zones: when imaging the heart chooses one focal zone at the region of interest to obtain better lateral resolution. (5) Dynamic range: narrow dynamic range provide better image as artifacts are eliminated. (6) High frame rate: high frame rate more than 25 frames per second. This can be achieved by narrowing the sector width and

reducing the depth. (7) Tint or color maps. Magnify the heart in order to fill 1/3rd to 1/4th of the ultrasound image. Insonate from the apical or right lateral aspect of the fetal chest when possible. Try to use cine loop to analyze cardiac and valves motion. Optimizing color Doppler for the fetal Echo: when examining fetal heart with color Doppler a rapid frame rate with an acceptable image quality is essential. The image quality can be improved by optimizing the use of the velocity, the wall filter, persistence, gain and color line density. Velocity scale or Pulse repetition frequency is used to determine the range of velocities in the region of interest. For AV valves, semilunar valves and the great vessels use high velocity range > 30 cm/sec. Pulmonary and systemic veins need low to mid velocity scale 10–20 cm/sec. Color filter: high filter is selected for AV valves, great vessels and low filters for pulmonary and systemic veins. Color gain-color gain should be initially set on low and gradually increases until the color information is optimized. Color Doppler image resolution and color line density: high color resolution is needed at periphery pulmonary vessels and fetal echo in early pregnancy. In this condition, the smallest color box is chosen to increase frame rate. In power, Doppler or HD mode reducing balance in combination with increasing gain can demonstrate clear delineation of flow events within the chambers and vessels. Near parallel insonation to the direction of blood flow helps to optimize image when color Doppler is used. Pulse Doppler allows Doppler wave forms quantifications for the assessment of cardiac abnormalities and functions.

Scanning technique: determine the fetal situs. The anatomic markers for visualization of four chamber view are: one complete rib on each side of fetal chest wall, the two inferior pulmonary veins noted along the posterior wall of the left atrium, the apex of the heart pointing to the left upper chest about 45° and descending Aorta in front and to the left of fetal spine. Slight tilt of the medial aspect of transducer from four chamber view plane, five chamber view (LVOT view) can be imaged. Sliding the transducer cranially (while maintaining the transverse orientation in the chest) from four chamber plane three vessel view can be obtained. The larger and left most vessel is main pulmonary artery, slightly smaller aorta in the middle and smaller SVC on right side. From three vessel plane cranial tilt of transducer the transverse view of arterial duct can be obtained. Still slight cranial tilt of transducer the transverse view of the aortic arch can be obtained. Anterior to the 3VT, there is hypoechoic structure which is slightly echogenic to the lungs is identified as thymus. Further cranial movement of the transducer with slightly oblique to the left from 3VT view, SVC connecting with brachiocephalic vein is noted. In this plane great vessels are not seen as they are inferior.

Four chamber view is basic plane for detail evaluation of the fetal heart. In this plane, look for situs, size, axis of the heart. In normal heart both atria equal in size, foramen ovale in midsection of atrial septum with leaflet of foramen ovale in left atrium. Tricuspid valve septal leaflet is more apically inserted on the septum than mitral valve. Both ventricles equal in size with intact ventricular septum and moderator band in right ventricle. Based on the position and fetal lie four types of four chamber views of fetal heart can be obtained: (1) When the fetal anterior chest wall is closest to the transducer the Apical four chamber view obtained (**Figure 4A**), in which the ultrasound beam is nearly parallel to ventricular septum. (2) When fetal right posterior chest wall is closest to the transducer, a basal four chamber view is obtained ultrasound beam is nearly parallel to ventricular septum. (3) When the fetal spine is neither anterior or posterior but closer to right or left lateral uterine walls a Lateral four chamber view is obtained (**Figure 4B**), where the ultrasound beam is perpendicular to inter ventricular septum. Apex of the heart, ventricles the atrioventricular valves, atrial, Ventricular longitudinal dimensions are optimally evaluated by apical four chamber view. Basal view allows optimum visualization

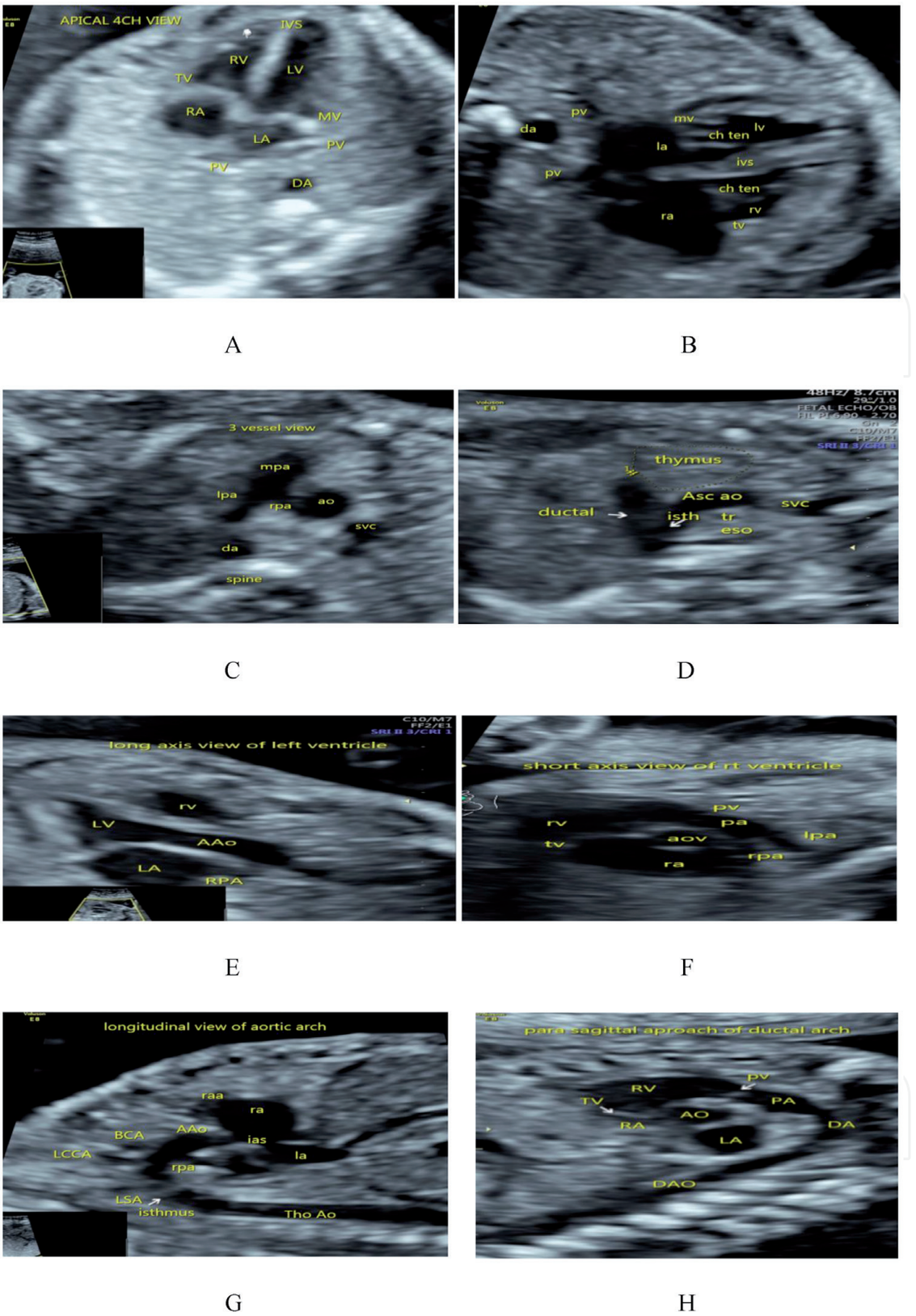


Figure 4.
(A) Apical 4ch view. (B) Lateral 4 ch view. (C) 3vv. (D) 3VT. (E) Long axis view of left ventricle. (F) Short axis view of right ventricle. (G) Longitudinal view of aortic arch. (H) Parasagittal approach of ductal arch.

of atria and atrioventricular valves. The lateral view allows adequate visualization of interventricular and interatrial septum, atrial and ventricular walls, ventricular contractility and septal thickness. From four chamber plane rotate the transducer 90° to get short axis views of the heart which provides a detail anatomic evaluation of spatial relationship of cardiac chambers and helps to evaluate ventricular size,

ventricular wall and septal thickness. Origin and relationship of the great vessels can also be evaluated. In most Apical short axis plane Posteromedial and anterolateral papillary muscle of left ventricle are imaged in same plane at 8- and 5-o clock positions. Basal or more posterior short axis plane demonstrates the muscular part of ventricular septum, Mitral and tricuspid valves.in this plane mitral valve is crescent shaped and has the appearance of fish mouth. Three vessel view/transverse pulmonary trunk view (**Figure 4C**) shows the large main pulmonary artery anteriorly, ascending aorta in middle and posteriorly placed small SVC. In this plane we can assess conotruncal abnormalities and abnormalities associated with vessel size, alignment, arrangement, number and location of descending aorta. Three VT view (**Figure 4D**) demonstrates transverse main pulmonary artery and ductal arch, Transverse Aortic arch and its isthmic region, and the cross section of SVC and trachea. Trachea in this plane appears as circular structure with echogenic wall and a black lumen. In normal 3VT plane aortic and ductal arches are located left of the spine and trachea and no vessel should be seen to the right of trachea. Color Doppler reveals anteroposterior flow in both arches. By reducing velocity scale svc and azygos arch blood flow entering into SVC can be recognized. Slightly more cranial to this plane the mammary arteries running on the lateral border of thymus can be visualized which is known as thymic box. More cranial plane will show blood flow in Left Brachiocephalic vein in left to right direction, always opposite to the direction of the Aortic flow. Abnormal findings in 3VT view are (1) Narrow or absent Aortic arch—which is suggestive for Tubular hypoplasia of aortic arch or aortic coarctation. (2) Narrow or absent pulmonary artery—can typically be seen in pulmonary stenosis or atresia showing decreased antegrade or revers flow in pulmonary artery. Common anomalies associated are Tetralogy of Fallot, Ebstein anomaly, some cases of Double outlet right ventricle, Tricuspid atresia with VSD. (3) Dilated transverse Aortic arch-Aortic valve stenosis with post stenotic dilatation of Aorta can be found as isolated finding. (4) Dilated pulmonary artery—a hugely dilated pulmonary artery typically found in Tetralogy of Fallot with absent pulmonary valve. Absent pulmonary valve syndrome is commonly in association with the absence of the arterial duct. (5) Only one normal sized great vessel suggests transposition, DORV, the visualized single vessel that is seen is Aorta with a posterior non visualized pulmonary artery. Aorta shows a typical right ward convex course. (6) Only one great vessel of larger size-single enlarged great vessel may be aortic arch in case of pulmonary atresia where pulmonary artery is small tortuous or even absent. On the other hand, it could be a dilated pulmonary artery in the presence of left outflow tract obstruction such as hypoplastic left heart syndrome where on color Doppler demonstrates reverse flow in aortic arch. In an interrupted aortic arch, the pulmonary artery is less dilated than in left outflow tract obstruction. In case of common arterial trunk, a single dilated great vessel is noted. (7) Course of Aortic arch to the right of the trachea–then it is known as Right sided aortic arch. It may be isolated or associated with left ductus arteriosus which forms a U configuration (U sign) with the trachea in the middle. Rarely both the arches may be right of the trachea forming right sided V sign. (8) Tortuous course of great vessel—most commonly found at the level of the ductal arch towards the end of gestation where it appears as S configuration. Sometimes it may also present in mid gestation. Tortuous ductal arch may be a normal variant with no clinical implications. An abnormal tortuous course of aortic arch may be seen in a cervical aortic arch where the aorta runs into the upper mediastinum which is distant from the course of pulmonary artery. (9) Connection between both great vessels—this may be found in Aortopulmonary window. It is a rare condition and may be found in tetralogy of Fallot or other conotruncal anomalies or an isolated finding. (10) Demonstration of four vessels—commonly present

in association with persistent left SVC. A double Aortic arch may also show four vessels, but it is generally detected by the presence of a right aortic arch. (11) Three vessels but no Right SVC – this condition may be seen with persistent left SVC. Dilated Superior vena cava: this may occur in case of Dilated Azygos vein in cases of interrupted IVC. This condition can also be depicted in the presence of supra cardiac type of total anomalous pulmonary venous drainage and also present in arteriovenous malformation of brain or vein of Galen aneurysm where increased venous drainage to SVC noted. In three VT view we can also asses the Thymus and left Brachiocephalic vein and also the other mediastinal structures like esophagus behind trachea, Bifurcation of trachea into both bronchi, the azygos vein connects to the SVC over the right bronchus referred as Azygos arch. In cases of conotruncal anomalies associated with hypoplastic or aplasia of thymus increases the suspicion for the presence of a 22q11 deletion.

Axial, oblique and sagittal views of great vessels (**Figure 3E–H**). The five chamber view displays ascending Aorta and demonstrates the left ventriculoarterial connection, the peri-membranous and muscular ventricular septa. Ascending aorta arises in between two AV valves in left to right orientation (directed toward right shoulder). There is a wide angle noted between the direction of ventricular septum and anterior wall of ascending Aorta which is absent in conotruncal anomalies where it is more parallel orientation with the septum. Fibrous–fibrous continuity of the posterior wall of the Aorta with the mitral valve and fibrous-muscular continuity of the anterior wall of Aorta and septum. This connection will be disrupted in Overriding of Aorta. At the level of five chamber view the two superior pulmonary veins enter the posterior walls of the left atrium. To obtain right ventricular outflow view (**Figure 4F**) from the mid sagittal plane of the chest by keeping the transducer in an oblique plane that is pointing from the right iliac bone to the left shoulder of the fetus. In this plane, right inflow and out flow tracts can be seen in same plane and are in perpendicular orientation. Left ventricle long axis view (**Figure 4E**)–to get this plane obtain sagittal view of fetal thoracic spine and slide the transducer from right to left parasagittal chest where we can image three planes one is superior and inferior venae cava second one is the aortic arch and lastly the ductal arch.

Longitudinal Aortic arch: by sliding the transducer to left parasagittal plane, we can obtain longitudinal aortic arch plane. In this view, aorta appears as a candy or a walking cane (**Figure 4G**) and arises centrally and superiorly in chest. Three arterial branches arise from superior aspect. Ductal arch view: it can be obtained by sliding the transducer still more left from the aortic arch view. Ductal arch (**Figure 4H**) arises from anterior aspect of chest with a wide angular curvature and is almost perpendicular to descending aorta and looks like hockey stick appearance. Ductal arch does not give any branches.

Normal variants in four Chamber view are (1) Echogenic intra cardiac focus (EIF) it may be single or multiple. In a patient at low risk of aneuploidy the presence of EIF after a normal detailed ultrasound should be considered as a normal variant; (2) pericardial fluid: if the pericardial fluid is less than 2 mm in thickness should be considered as normal variant; (3) mild ventricular disproportion in third trimester (left is smaller than right) can only be considered as normal variant after detailed ultrasound evaluation which ruled out coarctation of Aorta, TAPVC or other cardiac abnormalities associated with small left ventricle; (4) linear insertion of Atrioventricular valve could be found in normal fetuses and in fetuses with AVSD, depending on the plane in which the four chamber view is visualized (10). The area behind the 4ch view: during swallowing, esophagus may dilate and appears as a second vessel in front of Aorta but it will be decreased when swallowing ends.

Abnormal fetal echo: septal defects-Atrial septal defects-classified as Septum primum (ASD1), septum secundum (ASDII), Sinus venosus and coronary sinus defect. Septum secundum is the most common ASD which is around 80% of all ASDs and is mainly due to defect at foramen ovale. Next most common ASD is septum primum defect adjacent to both AV valves also known as partial AVSD. Sinus venosus ASD is noted posterior and superior to foramen ovale and inferior to the where IVC and **SVC** entering Right atrium. Coronary sinus ASD is rare and located at the site of the coronary sinus ostium in right atrium. Most ASDs cannot be diagnosed prenatally. ASD 1 can be identified as absent crux with linear insertion of both AV valves and Right to Left shunt noted. False positive diagnosis can be due to coronary sinus which mimics ASD1 where the flow is from left to right. Associated Cardiac and extra cardiac findings are AVSD, Anomalous pulmonary venous connections, single ventricle, Ebstein anomaly, tricuspid atresia and pulmonary atresia with intact ventricular septum. Venous abnormalities are common with sinus venosus defect (80–90%) and 10–15% with ASDII. ASD1 is associated with trisomy 21 and is also associated with Aortic coarctation. ASD secundum is associated with Holt-Oram syndrome.

Ventricular septal defect (VSD): these are the common congenital heart defects. VSDs are reported based on anatomic location on the septum. Most common VSD in neonates are perimembranous VSD (approximately 80%). Muscular, inlet and outlet VSDs are approximately reported in 5–20%, 5–8% and 5% respectively [11–13]. Whereas muscular VSDs (**Figure 5B**) are more common in prenatal period (aproxmately 80–90%) [14]. Small muscular VSDs are rarely diagnosed on gray scale color Doppler can be helpful in identifying the septal defect. Identification of VSDs is better in lateral chamber view. In Apical view drop out may mimic like VSD in such cases the borders of VSD appear echogenic. Most of the small muscular VSDs are bidirectional. On grayscale more commonly detected VSDs are perimembranous VSDs which are visualized in five chamber view where there is discontinuity between ventricular septum and medial wall of ascending Aorta. This type of VSDs is commonly associated with conotruncal abnormalities.

Atrioventricular septal defects (AVSD): common synonyms to AVSD are Endocardial cushion defect or AV canal defect. In the complete form of AVSD there is a combination of ASDI defect and VSD with common AV valve which usually have five leaflets (**Figure 5A**). Partial AVSD includes ASD1 and a cleft in mitral valve with separate AV valves but attaches at the same level. Transitional AVSD is a type of partial AVSD in this consists of two distinct AV annuli with large defect in atrial septum and small VSD.AVSDs can be classified as balanced or unbalanced.

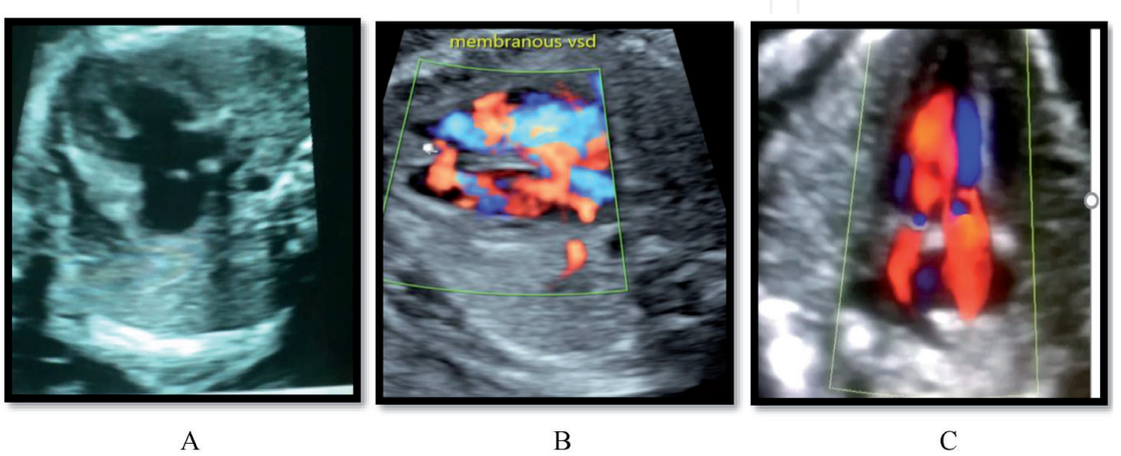


Figure 5.
(A) Atrioventricular septal defect. (B) Small membranous VSD. (C) Double inlet ventricle.

In unbalanced AVSDs the AV connections predominantly drains to one of the two ventricles results in ventricular disproportion and is typically found in Heterotaxy syndrome.

Incidence of AVSDs in infants with congenital heart diseases is 4–7.4% [15, 16]. Eighteen percent of prenatally detected CHDs are AVSDs [17]. It is best detected in Apical 4 chamber view. Ultrasound findings in AVSD in diastole are Absent Crux with a defect in the center of the heart noted. In systole the common AV valve is closed with loss of offset noted and appears like linear line across. Atrioventricular length ratio (AVL ratio) is increased in AVSD [10]. A cut off value for AVL ratio over 0.6 detects AVSD in 83% with false positive rate of 5.7% [18]. Presence of conotruncal Anomalies are more commonly associated with AVSD. Color and pulsed Doppler will demonstrate common regurgitation in complete AVSD. A regurgitant jet, which appears to arise from left ventricle should alert for the presence of a complete or partial AVSD as mitral regurgitation is rare in fetus (10). Associated cardiac findings are TOF, DORV, Right Aortic arch, aortic coarctation and other conotruncal anomalies. AVSD is also associated with Heterotaxy syndrome. Extra cardiac anomalies associated with AVSD are mainly trisomy 21 and also noted in trisomy 18 and 13. Antenatal diagnosis of AVSD, when isolated is associated with trisomy 21 in 58% of cases [19]. AVSD in combination of with hypoplastic left heart syndrome can be found in turners' syndrome (10). In a recent study 13% of CHARGE syndrome patients were associated with AVSDs [20]. AVSD may be recognized in early gestation at 11–13 weeks. Prenatal diagnosis of complete AVSD has low survival rate due to associated cardiac and extra cardiac findings whereas in isolated cases the outcome is excellent [21, 22].

Univentricular atrioventricular connection: this is due to failure of the development of bulboventricular loop stage. It describes a heart with one functioning ventricle with inflow from one or both atria. Synonymous terms for this condition are primitive ventricle, single ventricle, cor triloculare biatriatum, cor biloculare and double inlet ventricle (DIV). Three subgroups are identified in this condition, they are (1) double inlet (**Figure 5C**), where two atria connect to a single ventricle through two atrioventricular valves; (2) single inlet where one atrium connects to single ventricle through a single AV valve; (3) common inlet where both atria connected to single ventricle. Most common morphology of single ventricle is left ventricular morphology with right rudimentary chamber. Rarely right ventricular morphology with rudimentary left chamber or a ventricle of indeterminant morphology without rudimentary chamber can be seen. Cardiac anomalies which may show a single ventricle morphology are Hypoplastic left heart syndrome, Pulmonary atresia with intact septum, AVSD (large or unbalanced), single ventricle in Heterotaxy syndrome, corrected TGA with tricuspid atresia, mitral atresia with ventricular septal defect and more commonly found DIV is tricuspid atresia with ventricular septal defect. Double inlet left ventricle (DILV) presents as rudimentary right ventricle communicating with single ventricle with a VSD. Rudimentary right ventricle is a small outlet chamber. Great arteries usually arise in D or L malposition. The rudimentary outlet chamber in DILV is more commonly located on left side of the main ventricle (L looping) rarely on right side (D looping). Diagnosis is usually made on gray scale ultrasound. DIV with patent AV valves is well tolerated in fetus. Follow up ultrasound is important to look for outflow obstruction. Associated malformations are Atresia, hypoplasia or straddling of the atrioventricular valves, pulmonary outflow obstruction, subaortic outflow obstruction and conduction abnormalities.

Tricuspid Atresia with ventricular septal defect (TA): in this condition, the atrioventricular connection on right side is absent hence right ventricle is diminutive in size. Most of the cases tricuspid valve appears echogenic and thickened on

ultrasound. Aliasing noted across the mitral valve due to increased blood flow. Prenatal mitral regurgitation will have poor outcome. Inlet type of VSD typically perimembranous is present in almost all cases. The size of right ventricle depends on the size of VSD. Right ventricular contractility is normal with no myocardial thickening. A large interatrial connection in the form of large patent foramen ovale or atrial septal defect. There may be redundant flap of septum secundum that is bulging into Left atrium: malaligned interatrial and interventricular septa. The right ventricular outflow stenosis is a common finding in these cases. Pulmonary stenosis in this condition shows only decreased in arterial size and it is nonturbulent. Right aortic arch may be present. TA is classified into three types depends on spatial orientation of the great vessels. TA type 1 is a normally oriented great artery, which is most common (70–80%) (**Figure 6A**). Type 2 in 12–25% associated with D transposition of great vessels. Type3 is uncommon where there may be common arterial trunk or L Transposition. Four chamber view in TA is diagnostic. Ductal dependent pulmonary circulation in TA is seen in severe pulmonary stenosis or pulmonary atresia in association of small right ventricle. Doppler study of ductus venosus shows reverse A wave in end diastole most commonly noted at second or third trimester. Associated cardiac findings are great vessel stenosis/atresia, coarctation of aorta, interrupted aortic arch, persistent LSVC, Right aortic arch, pulmonary venous abnormality and juxtaposition of atrial appendage. Sometimes it may be associated with corrected transposition of great arteries where atretic tricuspid valve is on left side. Fetal karyotyping should be offered to rule out 22q11 microdeletion.

Ebstein anomaly, Tricuspid valve dysplasia and tricuspid regurgitation: in Ebstein anomaly (**Figure 6B**) septal and posterior leaflet of TV are displaced inferiorly from TV annulus towards the apex and originates from the right ventricular myocardium. Anterior leaflet maintains its normal attachment to annulus. Proximal portion of right ventricle is then in continuous with the right atrium and forms the atrialized portion of right ventricle. Gray scale ultrasound findings in four chamber view are cardiomegaly with increased cardiothoracic ratio. Progressive dilatation of right atrium with advancing gestation is noted. Attachment of septal leaflet of TV to the ventricular wall rather than valve annulus is noted by observing TV anatomy in systole and diastole by using cine loop which is essential to differentiating Ebstein anomaly from tricuspid valve dysplasia. In severe form of Ebstein anomaly with

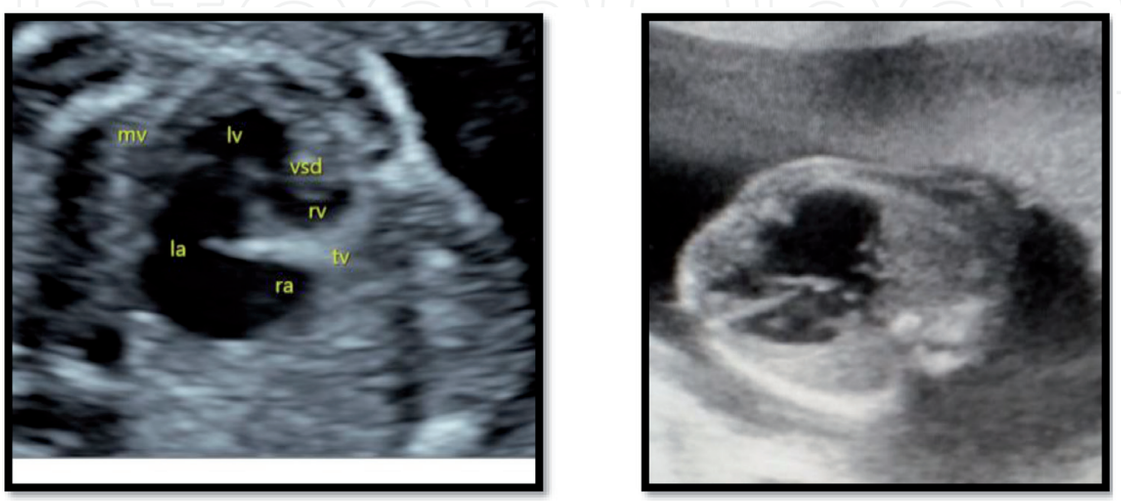


Figure 6.
(A) Tricuspid atresia with VSD. (B) Ebstein anomaly demonstrating apical displacement of tricuspid valve.

large atrialized ventricle there will be paradoxical movement of ventricular septum noted and pronounced cardiomegaly with compression of both lungs results in lung hypoplasia and may results in cardiac failure and hydrops. This anomaly may be associated with pulmonary stenosis or atresia. Color Doppler confirms the tricuspid regurgitation (TR). TR occurs during entire systole (holosystolic) with peak velocities of greater than 200 cm/sec. Systolic regurgitant jet typically arises from the middle of the right ventricle in contrast to other functional tricuspid regurgitations which arises at the level of annulus. Mild Ebstein cases may be missed in utero.

Tricuspid dysplasia: it involves the abnormalities of TV with normal insertion of leaflets. It also classified as a part of the pulmonary atresia with intact septum. Ultrasound findings are thickened leaflets which do not close properly in systole. Enlarged right atrium noted. It also associated with Right ventricular outflow obstruction and atrial septal defect. Color Doppler shows regurgitant jet arising from annulus. In the presence of significant right ventricular outflow obstruction the pulmonary artery may be of normal in size with minimal valvular excursion. Color Doppler will demonstrate reverse flow in the ductus arteriosus and may be of pulmonary valve regurgitation. Outcome of fetuses with this condition is good except in severe form it may be associated with heart failure, RVOT obstruction, severe tricuspid regurgitation and high rate of neonatal mortality.

Tricuspid regurgitation (TR): the regurgitant jet is limited to early or mid or whole systole is then referred as early systolic or mid systolic or holosystolic, respectively. Mild regurgitation peak systolic velocity varies from 30 to 70 cm/sec and in severe it may vary from 180 to 350 cm/sec. Mild TR is defined as the jet that extends less than 1/3rd of the distance to opposite atrial wall and covers an area less than 25% of right atrium. Trivial TR is common finding which is mild and nonholosystolic with maximum velocity less than 200 cm/sec. As Abuhamed et al, it is reported as 83% of fetuses in low risk in 14–16 weeks. In most of the cases it resolves in second trimester. TR is main component in Ebstein anomaly and TV dysplasia. RVOT obstructions such as pulmonary atresia with intact septum, pulmonary stenosis and constriction of ductus arteriosus are commonly associated with TR. It can also present in coarctation of aorta, hypoplastic left heart syndrome, Double outlet right ventricle and absent pulmonary valve syndrome. Volume overload like fetal anemia, Vein of Galen aneurysm, sacrococcygeal teratoma, chorioangioma, in the recipient twin in twin-twin transfusion syndrome and in fetal arrhythmias. Impairment of myocardial function like cardiomyopathies, sever IUGR, fetal hypoxia and infections or autoimmune myocarditis may associate with TR. In first trimester risk assessment TR is diagnosed when peak velocity of regurgitant jet is more than 60 cm/sec and duration involves at least half of the systole.

Hypoplastic left syndrome (HLHS): there are two main classic forms of HLHS noted. One involving both mitral and aortic atresia with severely hypoplastic left ventricle. The other form involves a small left ventricle with hyperechoic wall globular shape and poor contractility (**Figure 7A**). It is often difficult to differentiate HLHS from critical aortic stenosis on fetal echo. Ultrasound findings in four chamber view shows small hypokinetic left ventricle with apex is mostly formed by right ventricle. The size of left ventricle may be hypoplastic or normal in size or sometimes it may even dilate but always associated with poor contractility with decreased function this can be demonstrated by M mode. Aortic valve is atretic the mitral valve is patent but dysplastic and left ventricle is globular, hypokinetic with bright echogenic inner wall. Small left atrium with paradoxical movement of leaflet of foramen ovale. In five chamber view, aorta size will be less than 3 mm and at three vessel view dilated pulmonary trunk with small or nonvisible aorta noted. Color Doppler conforms retrograde flow from ductus arteriosus into aortic isthmus (**Figure 7B**). Pulse Doppler shows end diastolic pronounced reverse flow of A

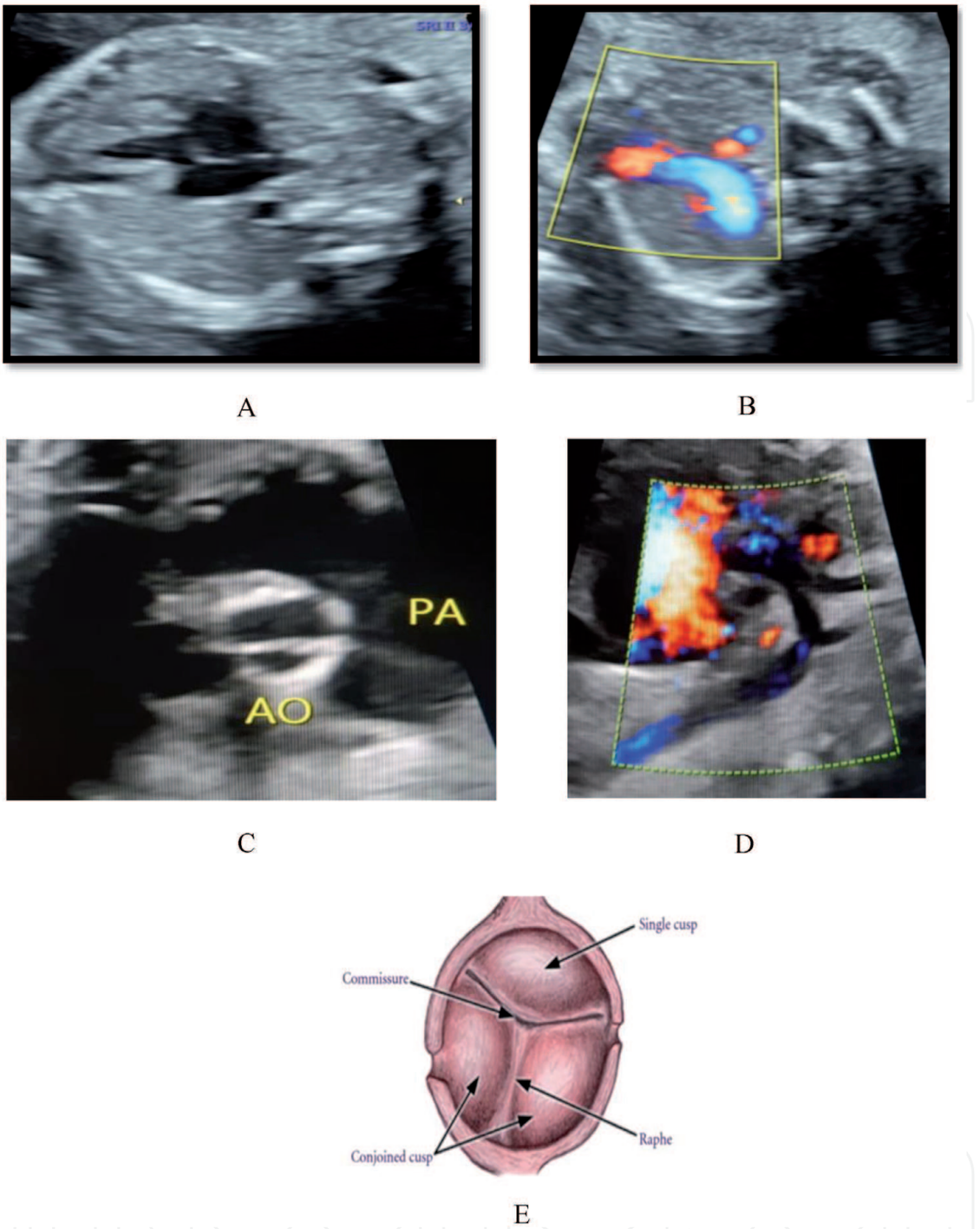


Figure 7.
(A) Hypoplastic left heart. (B) 3vessele view showing small aorta with reverse flow. (C, E) at the level of aortic root showing bicuspid aortic valve with single straight line across valve annulus. (D) Coarctation of aorta.

wave in pulmonary veins which is dependent on opening of foramen ovale size. In severe aortic stenosis left atrium may be dilated due to mitral valve regurgitation. Ascending aorta may show post stenotic dilatation. Peak velocities are generally >200 cm/sec. HLHS is associated with turner's syndrome, trisomies 13 and 18, Noonan syndrome, Smith-Lemli-Opitz syndrome and Holt-Oram syndrome. Follow up scans for every 4–6 weeks is recommended.

Aortic stenosis (AS) and Bicuspid aortic valve (BAV): it is classified as valvular, sub valvular and supra valvular depending on anatomical location of the obstruction in relation to aortic valve. Valvular AS is most diagnosed in prenatal ultrasound where the valve leaflets are dysplastic. It may be tricuspid with fused commissures,

bicuspid, unicuspid or noncommissural. Mild AS is difficult to diagnose prenatally. On gray scale aortic valve is thickened valve leaflets, doming of the cusps and lack of complete valve opening during systole noted. Doppler study shows turbulent antegrade flow through a normal-size or slightly dilated aortic arch. In shone complex the combination of left ventricular inflow and outflow obstruction with a normally contractile left ventricle. In shone complex there is narrowing of mitral valve orifices with a regular filling of small normally contractile left ventricle.

Bicuspid aortic valve: —this is an autosomal dominant inheritance pattern. It can also occur in other cardiac anomalies such as coarctation of Aorta, aortic stenosis and ventricular septal defects. [23]. In 80–90% cases BAV (**Figure 7C** and **E**) is made of two unequal leaflets in this larger one results from the fusion of the right and left cusps with central raphe and smaller leaflet which by itself is larger than one of the three leaflets of a normal aortic valve.

Valve leaflets are best visualized in third trimester and short axis view of aortic valve. Ultrasound findings in BAV are mildly dilated ascending aorta with increased flow velocities, echogenic aortic valve, visualization of valve leaflets in late systole.

Coarctation of Aorta (CoA) and interrupted aortic arch (IAA): —CoA involves narrowing of aorta at isthmic region typically located between left subclavian artery and ductus arteriosus (**Figure 7D**). If long segment of aorta involved, it is known as tubular hypoplasia. Recurrent rate for CoA is 2–6% for previously affected child 4% an affected mother [24, 25]. CoA is simple without associated with intra cardiac lesions and complex when it is associated with significant intra cardiac lesions like HLHS, Aortic atresia, unbalanced AV defect with a narrow-left ventricle, DORV, Tricuspid atresia with VSD and malposition of great vessels (type2), double inlet ventricle/single ventricle and corrected TGA. Ultrasound findings of CoA are ventricular disproportion left is <right. Ratio of right to left ventricular width should be more than 1.69 [26]. In this case, normal ventricular contractility and patent Mitral valve noted. Occasionally it may be associated with persistent left superior vena cava (PLSVC). In five chamber view normal ascending aorta normal or small aortic root especially if it is associated with perimembranous VSD or Aortic stenosis. On 3VT view narrowing of transverse aortic arch diameter more at isthmic region. PLSVC noted left of ductal arch. Narrowing and shelf is better assessed in longitudinal aortic arch section. Color Doppler confirms normal ventricular filling during diastole which differentiates CoA to HLHS. Narrowing of isthmus with normal flow without any aliasing noted. Look for shelf sign which is noted at the junction between the ductus arteriosus and descending aorta. An accurate diagnosis of CoA in early gestation is difficult as the ventricular disproportion found in early gestation may resolve with advancing gestation. Associated non cardiac findings are vascular anomalies like variations in brachiocephalic anatomy, berry aneurysm of circle of Willis. And nonvascular lesions involving multiple organs like Genitourinary, Musculoskeletal, Gastrointestinal and others may be present in 30% of children [27]. Associated aneuploidy rate is up to 35% (10), commonly associated with turner syndrome and occasionally with trisomy 13 and 18. CoA is difficult to detect prenatally with high false positive and false negative rates [28, 29].

Interrupted Aortic arch: it is classified into Type A, B and c in relation to brachiocephalic vessels. Type B is most common and is associated with large malaligned VSD and posterior displacement of infundibular septum. Type A is similar to Coarctation of aorta. Four chamber view usually appears on normal five chamber view shows large VSD with small aortic root showing nonturbulent flow. 3 VT view demonstrates absent continuity of aortic arch. Trachea appears to touch pulmonary artery due to absence of medially located aortic arch. It is associated with 22q11 deletion where the thymus is hypoplastic or absent in 3VT view.

Instead of candy cane appearance longitudinal aortic arch shows straight course with brachiocephalic and left common carotid vessels. Left subclavian artery arises from ductus arteriosus. Slightly dilated pulmonary trunk in 3 VT view. Occasionally associated with aberrant right subclavian artery (ARSA) or aberrant left subclavian artery (ALSA). 50% of IAA type B is associated with 22q11 microdeletion [30–32]. About 43% is associated with DiGeorge syndrome [33]. It may also associated with turner syndrome, pulmonary stenosis (PS), pulmonary atresia with intact ventricular septum (PA-IVS) and ductus arteriosus constriction.

Pulmonary stenosis: There may be infravalvular (infundibulum) narrowing or rarely supra valvular narrowing involving the main pulmonary artery or its branch. This is typically associated with tetralogy of fallot, Twin-twin transfusion syndrome. Valvular stenosis is due to fusion of valve commissures. Sometimes it may be due to unfused thickened dysplastic valve leaflets with associated pulmonary regurgitation usually associated with Noonan syndrome. Recurrence of PS is 2%in one affected sibling 6% when two siblings are affected. Ultrasound findings in PS in 4 chamber view are right ventricular hypertrophy with bulging of septum into left side TV shows normal leaflet excursion may associated with tricuspid regurgitation leads to right atrial dilatation. Right ventricular outflow view shows abnormal excursion, thickening and doming of valvular leaflets during systole. Valvular leaflets are visible within pulmonary artery throughout cardiac cycle. 3vv and 3vt view shows post stenotic dilatation. Color Doppler of PulmonaryValve shows turbulent antegrade flow with color aliasing. Pulse Doppler shows high flow velocities >200 cm/sec. Most of the cases shows antegrade flow in ductus arteriosus. Retrograde flow across the ductus arteriosus is a poor sign. In sever PS tricuspid valve shows holosystolic regurgitation. Ductus venosus shows reverse a wave. Associated cardiac lesions are tricuspid regurgitation, ASD, Aortic stenosis, Tricuspid Stenosis and Total anomalous pulmonary venous connection. Associated syndrome are Noonan, Beckwith-Wiedemann, Alagille and Williams-Beuren. Every 2–4 weeks of follow-up, scans are recommended.

Pulmonary atresia with intact ventricular septum (PA-IVS): —here, the PA is membranous type with complete fusion of commissures and normally developed infundibulum. In PA-IVS and hypoplastic right ventricle may show anomaly of coronary circulation known as ventriculocoronary arterial communication (VCAS). Systemic collateral arteries to lung are typical to PA-with VSD and not with PA-IVS. Ultrasound findings in PA-IVS (**Figure 8A** and **B**) are hypoplastic and hypokinetic right ventricle with thickened walls. The size of right ventricle May vary from nearly absent or hypoplastic or normal with poor contractility. Dysplastic tricuspid valve with narrow annulus and abnormal leaflet excursion noted. Hypoplastic main pulmonary artery noted. Color Doppler shows the lack of adequate filling of right ventricle during diastole. No antegrade flow across PV and reverse flow in ductus arteriosus. Dilated aorta with normal antegrade flow.in color Doppler VCACs which siphon right ventricular blood during systole, in the presence of Right ventricular cavity with diastolic filling and without significant regurgitation the apex or right ventricular wall as turbulent flow followed along the wall of right ventricle until the origin of the right or left coronary artery at the aortic root. Pulse Doppler demonstrates bidirectional turbulent flow with high velocities. Poor prognostic findings are severe tricuspid regurgitation, small tricuspid valve annulus (Z-score < 3or 4), small Rv/Lv length or width (<0.5), presence of VCAC, associated extracardiac abnormalities and chromosomal abnormalities.

Premature constriction of the ductus arteriosus: —DA is a muscular tube which connects the MPA at the origin of left PA to the descending aorta just distal to the left subclavian artery. It is the largest vessel in the fetus equal to that of descending aorta. In advanced gestation, increased deposition of collagen, elastin and

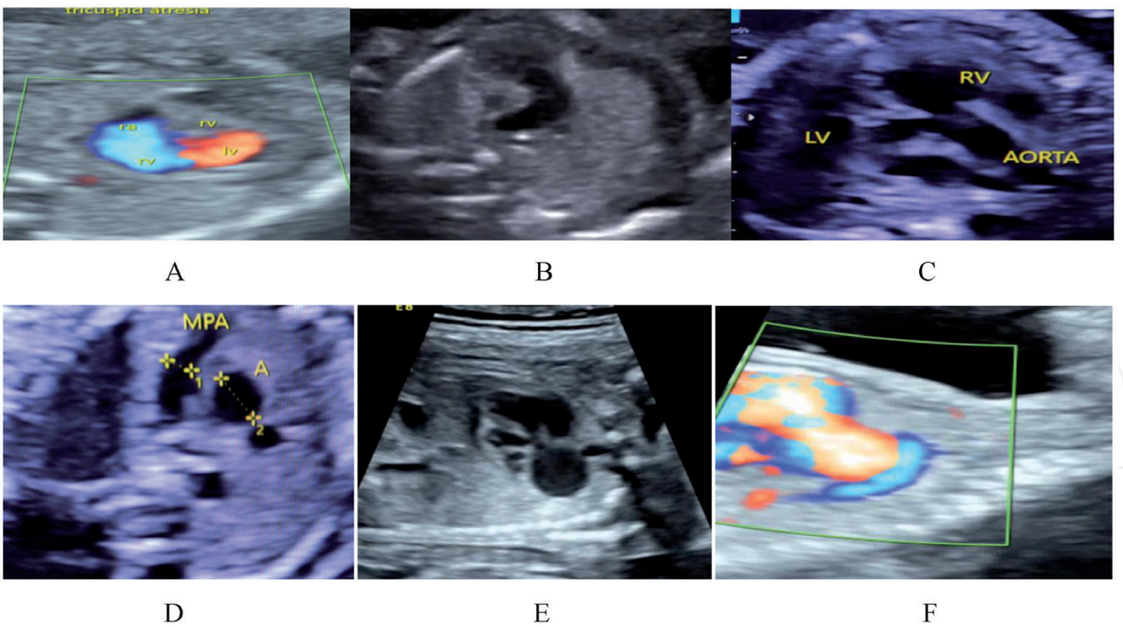


Figure 8.
(A) Pulmonary atresia with intact septum showing hypoplasia of right ventricle absence of blood flow across dysplastic tricuspid valve. (B) PA-IVS 3VT view showing small pulmonary artery. (C) Case of TOF in five chamber view showing large ventricular septal defect and dilated overriding aorta, with a parallel to the ventricular septum. (D) In TOF 3VV showing small pulmonary artery with dilated aorta and descending aorta on the right side of the spine as an associated with right aortic arch. (E, F) Absent pulmonary valve syndrome showing grossly dilated pulmonary artery showing high velocities with color aliasing.

glycoprotein with proliferation of smooth muscle to prepare for postnatal closure this narrowing can be monitored by prenatal ultrasound. Most cases of DA constriction are due to maternal drug therapy with prostaglandin synthetase inhibitors in late gestation. On gray scale it is difficult to diagnose this condition. Sometimes it may show dilated and hypokinetic right ventricle noted. Color Doppler reveals holosystolic tricuspid regurgitation. Dilated Right ventricular out flow tract, pulmonary artery and narrow ductus arteriosus by showing turbulent high diastolic and systolic velocities of around 200-300 cm/sec diastolic >35 cm/sec with PI less than 1.9. Interruption of drug therapy reverses the constriction within 24-48 hours but TR may persists longer time.

TOF (**Figure 8C** and **D**) is characterized by malaligned VSD (sub aortic), overriding aorta (aortic root overrides the VSD) assumes a parallel course to the septum, infundibular pulmonary stenosis and right ventricular hypertrophy. In 4chamber view normal or may show the VSD. Deviation of axis may be the first clue. On five chamber view shows perimembranous sub aortic VSD with overriding of aorta by demonstrating discontinuity between IVS and medial aortic wall which is known as malaligned VSD. There by the aorta is slightly shifted towards right side (Aortic dextroposition) so aorta receives blood from both right and left vetricles becomes dilated. Short axis or three vessel view shows narrow pulmonary artery. Mild TOF can be missed in second trimester scans due to subtle changes. Sometimes perimembranous VSD is evident without vessel diameter discrepancy in second trimester. Inflow of aorta shows aliased due to high perfusion where as in pulmonary artery may generally shows normal or only slightly increased flow. In TOF instead of PS, it may also associated with pulmonary atresia or absent pulmonary valve. Associated cardiac abnormalities are right sided aortic arches, sometimes TOF may associate with AVSD which increase the risk of chromosomal abnormalities [34]. A patent foramen ovale or atrial septal defect has been reported in 83% and PLSVC in 11%of new born with TOF [35]. Chromosomal abnormalities are around 30% with trisomies 21, 13 and 18 [20]. TOF may be associated with 22q11 microdeletion.

It may also found in Alagille syndrome, CHARGE syndrome [10]. Follow up scans to evaluate pulmonary artery growth and flow across the ductus arteriosus. A pulmonary valve and aortic valve ratio of<0.6 or a pulmonary valve Z-score of-3 at fetal fine echocardiogram (10) was highly sensitive but poor specific whereas classifying direction of flow in the ductus arteriosus as either normal or abnormal was both highly sensitive and specific for predicting the need for a neonatal intervention.

Pulmonary atresia with VSD: —includes atresia of PV hypoplasia of pulmonary artery and membranous or infundibular VSD, and an overriding of aorta. Distinct features that differentiate PA-VSD to TOF are no right ventricular outflow severe abnormalities of pulmonary circulation. The source of blood supply to lungs mainly depends on ductus arteriosus and systemic pulmonary collateral circulation or combination of both. Collaterals arising from descending aorta to lungs is known as major aortopulmonary collateral arteries (MAPCAs). This is commonly seen with diabetic mother.it is also commonly associated with 22q11 microdeletion. Ultrasound findings are similar to TOF except the diameter of aortic root has a larger diameter than TOF. In 3VV hypoplastic PA can occasionally visualized. In some cases, closed pulmonary valve with absent proximal pulmonary trunk noted. Ductus arteriosus tortuous and hypoplastic or may be dilated. Color Doppler demonstrates absent flow from right ventricle to pulmonary trunk and retrograde filling in branching pulmonary arteries. A longitudinal view of aorta from anterior or lateral approach may show the MAPCAs arising from descending aorta which need low velocity Doppler settings. Associated cardiac malformations are right sided aortic arch in 20–50% of cases [36]. ASD secundum type is commonly seen postnatally [37]. The absence of ductus arteriosus is in half the cases. MAPCAs are associated in about 44% of cases [38]. Extra cardiac association are with 22q11microdeletion which is found in 18–25%of fetuses [38, 39] previous type IV CAT where both the branching PA arising directly from descending aorta is currently classified as PA-VSD.

Absent pulmonary valve syndrome: absent or dysplastic or rudimentary pulmonary valve leaflets with an outlet VSD and an overriding Aorta noted in TOF-APVS. Most of the times it may be associated with absent patent ductus arteriosus. Branching arteries are grossly dilated and main pulmonary valve annulus shows signs of stenosis and severe insufficiency. It may also be associated with air way abnormalities. The rarer form of APVS with an intact septum and less dilated pulmonary trunk and a patent ductus arteriosus are reported. Four chamber may be normal or may show dilated right ventricle. Five chamber view may show VSD with overriding aorta with normal aortic root (not dilated as in TOF). Short axis and 3VT view shows aneurysmal dilatation of pulmonary artery and massively dilated branch pulmonary arteries.in most cases absent ductus arteriosus noted [40]. On color Doppler high velocities across PV annulus with typical to-and-fro flow with peak systolic velocities approx. Meas 200–250 cm/sec (**Figure 8E** and **F**). Sometimes there may be tricuspid regurgitation. A normal sized pulmonary artery may present in early gestation and full spectrum of APVS may not be evident till late gestation.so, diagnosis of APVS is difficult in early gestation. High mortality in APVS is due to cardiac failure or bronchomalacia.

Common arterial trunk (CAT): also known as persistent truncus arteriosus, aorticopulmonary trunk and truncus arteriosus communis. It is characterized by a single arterial trunk which arises from the base of the heart and gives origin to the systemic, pulmonary and coronary circulations. A large VSD is nearly absent in infundibular septum [41]. It is classified into four types. Type 1 is a short pulmonary trunk arises from the CAT and divides into right and left pulmonary arteries (**Figure 9A** and **B**). In type 2 and 3, both pulmonary arteries arise separately they may be close anatomically (from posteriorly) as in type2 or some distance from one another as in type3 (laterally) and in type 4 which is reclassified as PA-VSD.

The root is of CAT is large and has biventricular origin. In most of the cases root arise entirely from right ventricle rarely from left ventricle. On ultrasound levorotation of heart noted. Large malaligned VSD with overriding large vessel with color aliasing noted on 5 chamber view with pulmonary trunk arising from overriding vessel. Root is large with thickened dysplastic leaflets 3VT view show a single large vessel represents artic arch as the ductus arteriosus is not developed in more than 50% of cases [9]. In an interrupted aortic arch there is an absent continuity of aortic isthmus noted. Seventy percent of the cases aortic arch is left of trachea (right aortic arch). In about 1/3rd of cases hypoplastic or absent thymus noted [30, 38, 42]. Aortic arch abnormalities are commonly associated with CAT. 21–36% right sided aortic arch 15% with interrupted arch rarely hypoplasia of arch or double aortic arch noted. It may be associated with trisomies 21,18, 13 and 22q11 deletion is reported in 30–40%of the cases [43–45]. It is more commonly seen in diabetic mother.

Double outlet right ventricle (DORV): it is a type of ventriculoarterial connection, where both the great vessels arise from right ventricle. Several types of DORV (**Figure 9E** and **F**) noted, differing with regard to spatial relationship of the great arteries, location of the VSD, presence or absence of outflow obstruction. There are four types of anatomic relationships of the aorta to pulmonary artery at the level of semilunar valves [46]. They are right posterior aorta, a right anterior aorta, left anterior aorta and right lateral aorta. Location of VSD which is associated with DORV are sub aortic type, sub pulmonic type, doubly committed and remote. Ultrasound findings are abnormal ventriculoarterial connections. It may associate with atrioventricular valves like Mitral atresia, AVSD and double inlet ventricle. Five chamber view demonstrates VSD, the lack of continuity of the medial wall of the aorta with septum, origin of both vessels is from the anterior chamber. The location and relation of VSD to great arteries should be evaluated. Angle the transducer in between 5 chamber and 3VT view to demonstrate parallel course

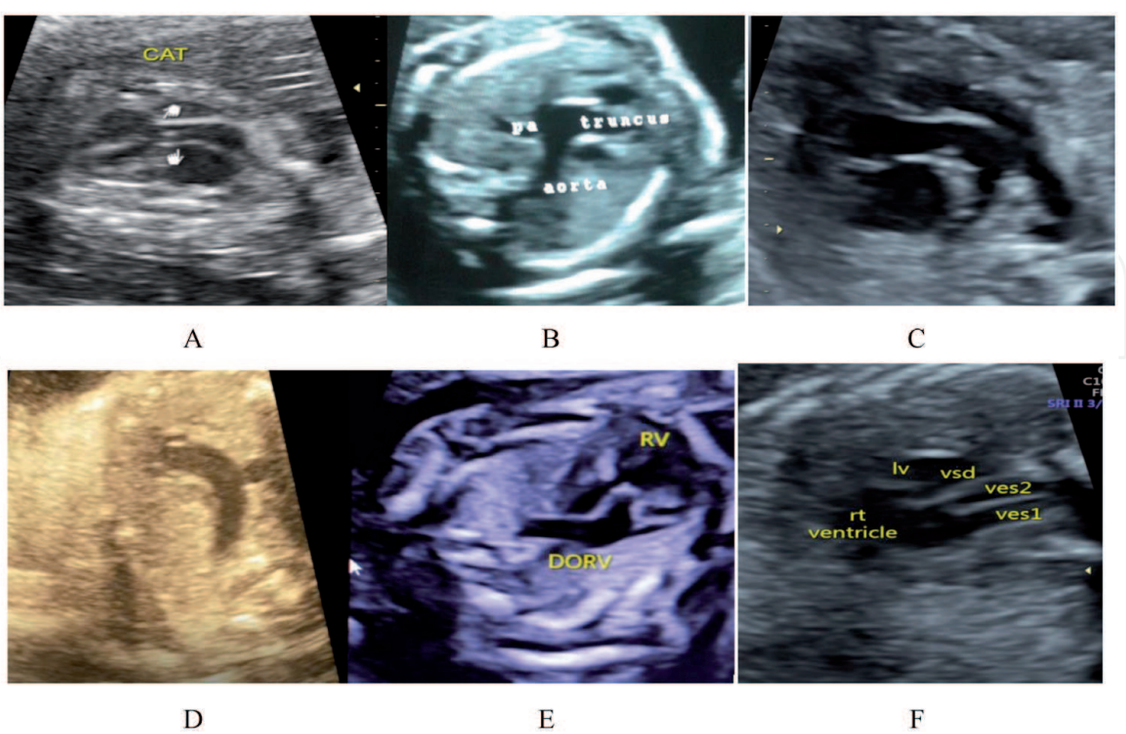


Figure 9.
(A) Five chamber view showing large overriding common arterial trunk. (B) Common arterial trunk with small pulmonary artery and large aorta. (C) Parallel origin of great vessels in DTGA. Aorta arising anteriorly from right ventricle and is right of the pulmonary artery which arises from left ventricle. (D) 3VT the aorta is seen as single large bent vessel as right convex shape i.e., I sign. (E, F) outflow tract view showing DORV.

of great vessels. Pulmonary artery atresia/stenosis is more common than aortic involvement. PS is most commonly associated with malformation in about 70% of cases [47] right sided aortic arch may be seen on 3VT view. The abnormal anatomic relation of great arteries can be better depicted on color Doppler. If atresia/stenosis of vessel is present, color Doppler helps to demonstrate a single large vessel on 3VT view. Cardiac malformations associated with DORV are mitral atresia, cleft in anterior mitral leaflet, ASD, AVSD, sub aortic stenosis, aortic coarctation, right aortic arch, persistent left svc and anomalous pulmonary venous return. DORV with AVSD may be noted in isomerism. DORV may also be noted with complex corrected transposition. In 12–40% of cases, chromosomal abnormalities including trisomies 18, 13 and 22q11 microdeletion are noted [48–50]. Poor prognostic signs associated with DORV are tubular or hypoplastic aorta, pulmonary atresia, hypoplastic left ventricle/single ventricle physiology, mitral atresia, AVSD and situs ambiguous.

Complete and congenitally corrected Transposition of great arteries: in complete transposition of great arteries (DTGA) atrioventricular concordance and ventriculoarterial discordance. Both great arteries display parallel course with aorta anterior and to right of pulmonary artery. It may present as simple or associate with other cardiac malformations (complex DTGA). VSD and PS are common association with DTGA and may be present alone or in combination in up to 30–40% of cases. Ultrasound findings are that five chamber view shows pulmonary artery arising from left ventricle and bifurcating shortly from its origin and aorta is arising from right ventricle in an anterior and parallel course to pulmonary artery. Transverse view in upper thorax demonstrates a single large anteriorly and superiorly placed vessel with SVC to its right. The single large vessel noted in 3VT view is Aorta and it assumes a right convex shape known as I sign (**Figure 9C** and **D**). On longitudinal view hockey stick appearance to aorta and candy cane appearance to pulmonary artery noted. Congenitally corrected transposition of great vessels (LTGA/ccTGA): characterized by atrioventricular and ventriculoarterial discordance. In ccTGA aorta is located anteriorly and left of the pulmonary artery. It has 2% recurrence risk in first degree relatives [51]. In some cases of ccTGA with dextrocardia or situs inversus, right atrium may be left sided and with double discordance of the ventricles and great vessels hemodynamically this condition resembles D-TGA. Situs inversus with ccTGA is noted in 5% of cases and dextro or mesocardia in 25% cases [52]. Sequential segmental analysis of fetal heart should be done to rule out situs abnormalities. Four chamber view assess ventricular morphology. Morphologic right ventricle in ccTGA is left posterior and is connected to left atrium. Morphology of right ventricle such as moderator band, apical attachment of AV valve, irregularity of the endocardial surface and a more triangle shape. Morphologic left ventricle is in right anterior and is connected to right atrium where it is characterized by smooth surface and elongated appearance with an Apex formation. Out flow tract shows pulmonary artery arising from right sided morphologic left ventricle and aorta from left sided right ventricle in parallel course with aorta anterior and left of the pulmonary artery. Here the PA arising from right sided ventricle has a course towards left side and aorta arising from left sided ventricle also courses towards left where as in DTGA aorta is coursing towards right side. Poor long-term prognosis is noted in ccTGA in association with Ebstein malformation of tricuspid valve, degree of TR, systemic right ventricular dysfunction and complete heart block. Survival rates exceeded 80% in prenatally diagnosed ccTGA when pregnancy was continued [53, 54].

Right aortic arch, double aortic arch and aberrant subclavian artery (ARSA):— Ultrasound findings in right aortic arch are: four chamber view shows descending aorta and is more centrally located. The detection and classification can be

achieved in 3VT view where the aorta courses to right side of trachea. There are three subgroups: (1) right aortic arch with right ductus arteriosus (Rt V sign) here both the aorta and ductus arteriosus merges together on the right of trachea with a V configuration. There is mirror imaging branch pattern to normal left aortic arch noted. (2) Right aortic arch with left ductus arch (Usign): in this pattern, right-sided aorta with DA on to the left of trachea. Trachea is in between the DA and aorta (**Figure 10A** and **C**) in most of the cases it is associated with aberrant left subclavian artery. Arising from junction between ductus arteriosus and descending aorta called Kommerell diverticulum [55]. (3) Double aortic arch-aorta courses to right side of the trachea but bifurcates and one goes to left another one to right of trachea resembling Lambda configuration. Esophagus and trachea is entraped. Two vessels arise from each aortic arch.

Aberrant right subclavian artery (ARSA): in this, instead of three vessels, four vessels arises from left sided aortic arch from proximal to distal are right common carotid, left common carotid, left subclavian and ARSA. ARSA (**Figure 10B**) arises distally from aortic arch and courses towards left side of chest behind trachea and esophagus and to the right upper arm. This is also known as retrophayngeal right sub clavian artery or Lusorian artery. On 3VT ARSA is noted at the junction of aortic arch and ductus arteriosus with a course behind the trachea towards right shoulder. It may be isolated or commonly associated with conotruncal anomaly, 22q11microdeletion and trisomy 21.

Heterotaxy and situs inversus:-normal development and positioning of thoracic and abdominal organs is situs solitus. Situs inversus is mirror image arrangement of thoracic and abdominal viscera. Different arrangement of thoracic and abdominal viscera other than situs solitus and situs inversus is situs ambiguous or Heterotaxy syndrome or isomerism is characterized by an abnormal symmetry of the viscera that are normally dissimilar, which has different arrangement than situs solitus or situs inversus. This is due to abnormal lateralization of thoracic and abdominal viscera and is frequently associated with complex cardiac anomalies. Isomerism may be of right or left. Left isomerism (**Figure 11A**) the Ultrasound finding that strongly suggested in the presence of a combination of at least two of the following: (1) Viscerocardiac heterotaxy; (2) complete AVSD or other structural heart disease; (3) interruption of IVC with azygos continuation; (4) early fetal heart block; Right isomerism (**Figure 11B** and **C**) should be suspected in the presence of a combination of at least two of the following: (1) Viscerocardiac Heterotaxy, (2) structural heart disease, namely complete AVSD; (3) juxtaposition of IVC and descending aorta. If both interrupted IVC and complete heart block are observed, we can almost be sure that there is LAI. Left isomerism: bilateral left sidedness with maldevelopment of right-side structures. Morphologic left characteristics such as bilateral left bronchi, bilobed lungs, finger-like atrial appendages. Inferior vena

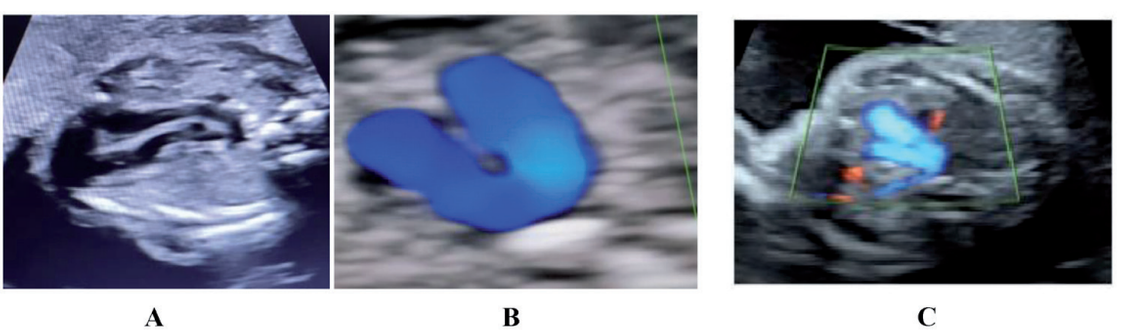
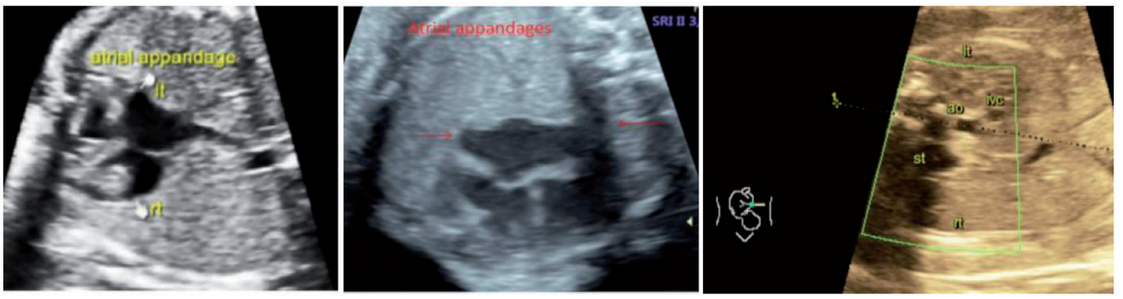


Figure 10.
(A) CCTGA with right aortic arch. (B) Right aortic arch with Usign. (C) ARSA coursing behind the trachea.



A B C

Figure 11.
(A) Left isomerism: finger like narrow bilateral symmetric left atrial appendages. (B) Broad pyramidal shaped symmetrical right atrial appendages in right isomerism. (C) Juxtaposition of aorta and IVC. Both on left side.

caval (IVC) interruption with azygous continuation, in which the hepatic segment of the inferior cava is absent, and the distal segment continues with the azygos or hemiazygos vein. A parasagittal view of abdomen and chest can demonstrate the azygose vein posterior to descending aorta. Color Doppler study shows opposite direction of flow in aorta and azygose vein noted. Right isomerism: bilateral structures with morphologic right characteristics, such as bilateral right atria, broad triangular atrial appendages, bilateral eparterial bronchi, and trilobed lungs. Ultrasound findings in right isomerism are Dextroversion is more common than left isomerism. More common cardiac anomaly associated are unbalanced AVSD and most of the times univentricular atrioventricular connection. TGA and DORV associated with pulmonary stenosis or atresia are associated commonly with right isomerism. Supracardiac TAPVC associated with right isomerism in 30%, infra diaphragmatic in 25% and cardiac in 30% and mixed in 15% observed. Visceral anomalies like malrotation and malfixation of the bowel, preduodenal portal vein, gastric volvulus, esophageal hiatal hernia, and biliary atresia are common in both left and right isomerism.

Situs inversus: in this, there will be mirror image arrangement of visceral organs in abdomen and thorax. Situs inversus associated cardiac anomalies are 0.3–5%in fetuses and newborn [56]. About 50% patients of Kartagener syndrome have situs inversus [57]. In situs inversus liver, IVC are on left side and spleen, stomach, heart and aorta are on right side.

Anomalies of systemic and pulmonary venous connections: they can occur as isolated or may be associated with simple cardiac anomalies like ASD and complex like Heterotaxy syndrome. Systemic venous malformation involves anomalies of IVC, SVC and coronary sinus. Persistent Left SVC (PLSVC) and interrupted IVC with azygos continuation are the most common systemic venous anomalies. The other rare venous malformation is abnormalities of ductus venosus, hepatic or umbilical veins.

PLSVC: it joins the coronary sinus on posterior left atrioventricular valve and drains to right atrium. On fetal echocardiography PLSVC (**Figure 12A**) appears as an extraneous vessel noted left to the ductal arch in three vessel trachea view. Very rarely PLSVC may be seen with absent right SVC (**Figure 12B**). Isolated PLSVC usually has no clinical significance. It is more commonly associated with other cardiac conditions (23% of PLSVC cases) They are Atrioventricular septal defect (AVSD), Double outlet right ventricle (DORV), Left out flow tract obstructive anomalies, Conotruncal anomalies and ventricular septal defect. The other Extra cardiac malformations including Heterotaxy syndrome (41–45%), Esophageal atresia, diaphragmatic hernia, IVC malformations, complex malformation syndromes

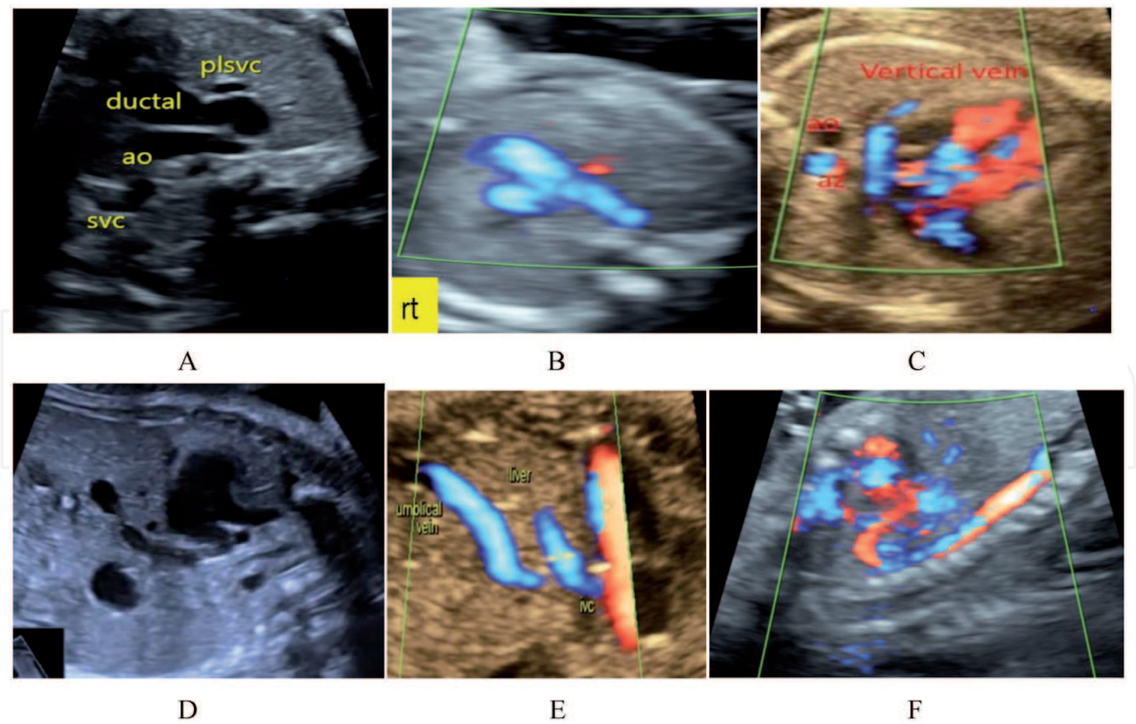


Figure 12.
(A) 3VTshowing 4vessels as in double SVC. (B) 3VT view showing 3 vessels in PLSVC with absent right SVC. (C) Supracardiac type of TAPVC. (D) Infra cardiac type of TAPVC with vertical vein coursing downwards. (E) Interrupted IVC-absent hepatic portion of IVC. (F) Azygos arch.

and chromosomal anomalies. Plane slightly inferior to four chamber view dilated coronary sinus can be noted. Normal diameter of coronary sinus is 1–3 mm; it courses perpendicular to inter atrial septum and opens into posterior wall of right atrium.

Interrupted IVC with azygos continuation: this is due to failure to form right subcardinal hepatic anastomosis results in absent hepatic segment of IVC (**Figure 12E** and **F**). The sonographic landmarks are dilated azygos vein alongside the Aorta in Abdominal circumference plane and four chamber view plane. In sagittal abdominal plane the descending Aorta and azygos vein run side by side with opposite direction of flow. Interrupted IVC has also been reported in isolated entity. In such cases it is clinically silent. It is more commonly associated about 80–90% with Left atrial isomerism.

Total Anomalous pulmonary connections (TAPVC) and partial anomalous connections (PAPVC): four pulmonary veins (PV) superior and inferior on both sides drains into posterior wall of left atrium. TAPVC involves all the pulmonary vein drains into right atrium directly or indirectly. Depending upon the anatomic site of anomalous connection four types of TAPVC noted 1supracardiac which is most common type, 2 cardiac, 3infra cardiac and mixed type [33]. Diagnosis is difficult prenatally. In a large series involving 424 cases with TAPVC only 8 were diagnosed prenatally. Ultrasound findings in four chamber view shows disproportion of right and left cardiac sizes with enlarged right sided chambers due to venous return. Area behind the heart in four chamber view plane shows increased distance between descending aorta and posterior wall of left atrium. There is no connection between pulmonary veins and left atrium. Which drains into the confluent vein or vertical vein. The confluent vein courses upward towards upper thorax drains into brachiocephalic vein in turn drains into Right atrium though SVC (**Figure 12C**). Vertical vein coursing downwards across the diaphragm in infra cardiac type (**Figure 12D**) or directly connects to right atrium or coronary sinus in cardiac type. In supra

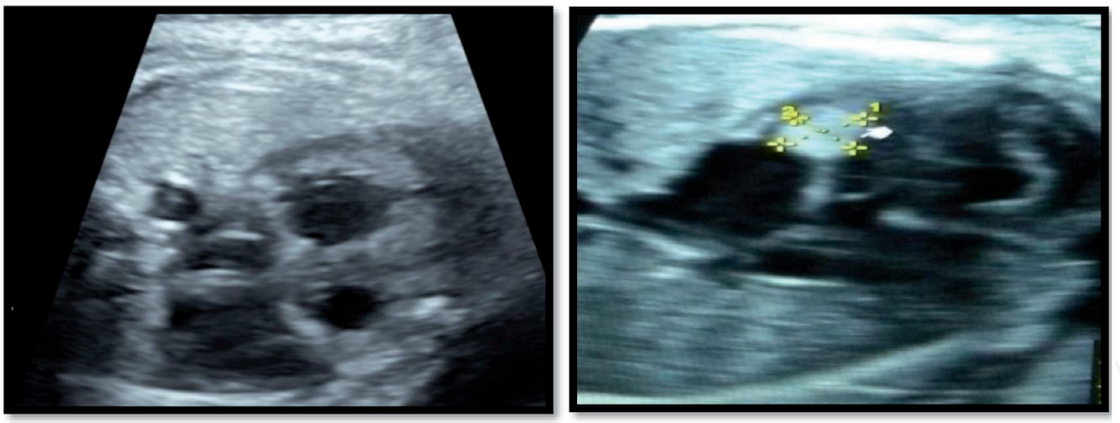


Figure 13.
(A) Hypertrophic cardiomyopathy. (B) Rhabdomyoma.

cardiac TAPVC at 3 vessel trachea view plane a fourth vessel i.e. vertical vein noted left of ductal arch. Blood flow in vertical vein in supra cardiac type flows superiorly towards upper thorax and there is dilated brachiocephalic vein noted. In cardiac type dilated coronary sinus in the absence of PLSVC should raise the suspicion of cardiac TAPVC. Infracardiac TAPVC the vertical vein formed behind the left ATRIA courses downwards along with esophagus through the diaphragm and drains into. portal veins. It is very difficult to see on routine gray scale ultrasound.in PAPVC difficult to detect and it is rarely reported prenatally. Scimitar syndrome: it is a combination of right lung hypoplasia, right pulmonary artery hypoplasia and PAPVC. In four chamber view, dextrocardia with right lung hypoplasia is noted. Right inferior pulmonary vein drains into IVC instead of left atrium.

Cardiomyopathies and fetal heart tumors: it is a disease of myocardium and is commonly associated with abnormal cardiac function. This can be manifested as dilated cardiomyopathy showing enlarged heart with dilated and decreased contractility or hypertrophic cardiomyopathy (**Figure 13A**) showing enlarged heart in association with ventricular wall hypertrophy. There may be reduced lumen of the effected ventricle. Mostly associated with diabetes mellitus. Pericardiac effusion is seen in cardiomyopathy. Heart tumors: 80–90% are rhabdomyomas (**Figure 13B**) but can also be teratoma, fibroma, myxoma, hamartoma, rhabdomyosarcoma and others. In Rhabdomyoma demonstrated in ultrasound as Ovale or circular well-defined echogenic mass. It may occur in the septum, wall or may be in the atrium and commonly associated with tuberous sclerosis.

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References

[1] Hoffman JI, Christianson R. Congenital heart disease in a cohort of 19,502 births with long-term follow-up. The American Journal of Cardiology. 1978;**42**:641-647

[2] Benson DW. The genetics of congenital heart disease: A point in the revolution. Cardiology Clinics. 2002;**20**:385-394

[3] Fogel M, Copel JA, Cullen MT, et al. Congenital heart disease and fetal thoracoabdominal anomalies: Associations in utero and the importance of cytogenetic analysis. American Journal of Perinatology. 1991;**8**:411-416

[4] Friedman AH, Copel JA, Kleinman CS. Fetal echocardiography and fetal cardiology: Indications, diagnosis and management. Seminars in Perinatology. 1993;**17**:76-88

[5] Crawford DC, Chita SK, Allan LD. Prenatal detection of congenital heart disease: Factors affecting obstetric management and survival. American Journal of Obstetrics and Gynecology. 1988;**159**:352-356

[6] Bronshtein M, Gover A, Zimmer EZ. Sonographic definition of the fetal situs. Obstetrics and Gynecology. 2002;**99**:1129-1130

[7] Salomon LJ, Baumann C, Delezoide AL, et al. Abnormal abdominal situs: What and how should we look for? Prenatal Diagnosis. 2006;**26**:282-285

[8] Comstock CH. Normal fetal heart axis and position. Obstetrics and Gynecology. 1987;**70**:255-259

[9] Smith RS, Comstock CH, Kirk JS, et al. Ultrasonographic left cardiac axis deviation: A marker for fetal anomalies. Obstetrics and Gynecology. 1995;**85**:187-191

[10] Abuhamad AZ, Chaoui R. A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts. Lippincott Williams & Wilkins; 2012

[11] Rubio AE, Lewin MB. Ventricular septal defects. In: Allen HD, Driscoll DJ, Shaddy RE, et al, editors. Moss and Adams' Heart Disease in Infants, Children, and Adolescents. 8th ed. Baltimore, MD: Williams & Wilkins; 2012. pp. 713-721

[12] Lincoln C, Jamieson S, Joseph M, et al. Transatrial repair of ventricular septal defects with reference to their anatomic classification. The Journal of Thoracic and Cardiovascular Surgery. 1977;**74**:183-190

[13] Soto B, Becker AE, Moulaert AJ, et al. Classification of ventricular septal defects. British Heart Journal. 1980;**43**:332-343

[14] Gomez O, Martinez JM, Olivella A, et al. Isolated ventricular septal defects in the era of advanced fetal echocardiography: Risk of chromosomal anomalies and spontaneous closure rate from diagnosis to age of 1 year. Ultrasound in Obstetrics & Gynecology. 2014;**43**:65-71

[15] Ferencz C, Rubin JD, Loffredo CA, et al. Epidemiology of congenital heart disease: The Baltimore-Washington infant study. In: Perspectives in Pediatric Cardiology. Mount Kisko, NY: Futura Publishing; 1981-1989. p. 1993

[16] Cetta F, Minich LL, Maleszewski JJ, et al. Atrioventricular septal defects. In: Allen HD, Driscoll DJ, Shaddy RE, et al, editors. Moss and Adams' Heart Disease in Infants, Children, and Adolescents.

8th ed. Baltimore, MD: Williams & Wilkins; 2012. pp. 691-712

[17] Allan LD, Sharland GK, Milburn A, et al. Prospective diagnosis of 1,006 consecutive cases of congenital heart disease in the fetus. Journal of the American College of Cardiology. 1994;**23**:1452-1458

[18] Machlitt A, Heling KS, Chaoui R. Increased cardiac atrial-to-ventricular length ratio in the fetal four-chamber view: A new marker for atrioventricular septal defects. Ultrasound in Obstetrics & Gynecology. 2004;**24**:618-622

[19] Delisle MF, Sandor GG, Tessier F, et al. Outcome of fetuses diagnosed with atrioventricular septal defect. Obstetrics and Gynecology. 1999;**94**:763-767

[20] Corsten-Janssen N, Kerstjens-Frederikse WS, du MarchieSarvaas GJ, et al. The cardiac phenotype in patients with a CHD7 mutation. Circulation. Cardiovascular Genetics. 2013;**6**:248-254

[21] Aubert S, Henaine R, Raisky O, et al. Atypical forms of isolated partial atrioventricular septal defect increase the risk of initial valve replacement and reoperation. European Journal of Cardio-Thoracic Surgery. 2005;**28**:223-228

[22] Studer M, Blackstone EH, Kirklin JW, et al. Determinants of early and late results of repair of atrioventricular septal (canal) defects. The Journal of Thoracic and Cardiovascular Surgery. 1982;**84**:523-542

[23] Allan LD, Crawford DC, Chita SK, et al. Familial recurrence of congenital heart disease in a prospective series of mothers referred for fetal echocardiography. The American Journal of Cardiology. 1986;**58**:334-337

[24] Nora JJ, Berg K, Nora AH. Cardiovascular Diseases: Genetics, Epidemiology, and Prevention. New York, NY: Oxford University Press; 1991

[25] Hornberger LK, Sahn DJ, Kleinman CS, et al. Antenatal diagnosis of coarctation of the aorta: A multicenter experience. Journal of the American College of Cardiology. 1994;**23**:417-423

[26] Paladini D, Volpe P, Russo MG, et al. Aortic coarctation: Prognostic indicators of survival in the fetus. Heart. 2004;**90**:1348-1349

[27] Brown DL, Durfee SM, Hornberger LK. Ventricular discrepancy as a sonographic sign of coarctation of the fetal aorta: How reliable is it? Journal of Ultrasound in Medicine. 1997;**16**:95-99

[28] Sharland GK, Chan KY, Allan LD. Coarctation of the aorta: Difficulties in prenatal diagnosis. British Heart Journal. 1994;**71**:70-75

[29] Chaoui R, Heling KS, Lopez AS, et al. The thymic-thoracic ratio in fetal heart defects: A simple way to identify fetuses at high risk for microdeletion 22q11. Ultrasound in Obstetrics & Gynecology. 2011;**37**:397-403

[30] Chaoui R, Kalache KD, Heling KS, et al. Absent or hypoplastic thymus on ultrasound: A marker for deletion 22q11.2 in fetal cardiac defects. Ultrasound in Obstetrics & Gynecology. 2002;**20**:546-552

[31] Volpe P, Marasini M, Caruso G, et al. Prenatal diagnosis of interruption of the aortic arch and its association with deletion of chromosome 22q11. Ultrasound in Obstetrics & Gynecology. 2002;**20**:327-331

[32] Van Mierop LH, Kutsche LM. Cardiovascular anomalies in DiGeorge

syndrome and importance of neural crest as a possible pathogenetic factor. The American Journal of Cardiology. 1986;**58**:133-137

[33] Uretzky G, Puga FJ, Danielson GK, et al. Complete atrioventricular canal associated with tetralogy of Fallot. Morphologic and surgical considerations. The Journal of Thoracic and Cardiovascular Surgery. 1984;**87**:756-766

[34] Rao BN, Anderson RC, Edwards JE. Anatomic variations in the tetralogy of Fallot. American Heart Journal. 1971;**81**:361-371

[35] Poon LC, Huggon IC, Zidere V, et al. Tetralogy of Fallot in the fetus in the current era. Ultrasound in Obstetrics & Gynecology. 2007;**29**:625-627

[36] Bharati S, Paul MH, Idriss FS, et al. The surgical anatomy of pulmonary atresia with ventricular septal defect: Pseudotruncus. The Journal of Thoracic and Cardiovascular Surgery. 1975;**69**:713-721

[37] Vesel S, Rollings S, Jones A, et al. Prenatally diagnosed pulmonary atresia with ventricular septal defect: Echocardiography, genetics, associated anomalies and outcome. Heart. 2006;**92**:1501-1505

[38] Boudjemline Y, Fermont L, Le Bidois J, et al. Prevalence of 22q11 deletion in fetuses with conotruncal cardiac defects: A 6-year prospective study. The Journal of Pediatrics. 2001;**138**:520-524

[39] Wertaschnigg D, Jaeggi M, Chitayat D, et al. Prenatal diagnosis and outcome of absent pulmonary valve syndrome: Contemporary single-center experience and review of the literature. Ultrasound in Obstetrics & Gynecology. 2013;**41**:162-167

[40] Sharland G. Common arterial trunk. In: Allan LD, Hornberger LK, Sharland GK, editors. Textbook of Fetal Cardiology. London, England: Greenwich Medical Media; 2000. pp. 288-303

[41] Volpe P, Paladini D, Marasini M, et al. Common arterial trunk in the fetus: Characteristics, associations, and outcome in a multicentre series of 23 cases. Heart. 2003;**89**:1437-1441

[42] Machlitt A, Tennstedt C, Korner H, et al. Prenatal diagnosis of 22q11 microdeletion in an early second-trimesterfetus with conotruncal anomaly presenting with increased nuchal translucency and bilateral intracardiac echogenic foci. Ultrasound in Obstetrics & Gynecology. 2002;**19**:510-513

[43] Sridaromont S, Feldt RH, Ritter DG, et al. Double outlet right ventricle: Hemodynamic and anatomic correlations. The American Journal of Cardiology. 1976;**38**:85-94

[44] Bradley TJ, Karamlou T, Kulik A, et al. Determinants of repair type, reintervention, and mortality in 393 children with double-outlet right ventricle. The Journal of Thoracic and Cardiovascular Surgery. 2007;**134** 967-973.e6

[45] Obler D, Juraszek AL, Smoot LB, et al. Double outlet right ventricle: Aetiologies and associations. Journal of Medical Genetics. 2008;**45**:481-497

[46] Chaoui R, Korner H, Bommer C, et al. Prenatal diagnosis of heart defects and associated chromosomal aberrations [in German]. Ultraschall in der Medizin. 1999;**20**:177-184

[47] Becker TA, Van Amber R, Moller JH, et al. Occurrence of cardiac malformations in relatives of children with transposition of the great arteries. American Journal of Medical Genetics. 1996;**66**:28-32

[48] Atallah J, Rutledge JM, Dyck JD. Congenitally corrected transposition of the great arteries (atrioventricular and ventriculoarterial discordance). In: Allen HD, Driscoll DJ, Shaddy RE, et al, editors. Moss and Adams' Heart Disease in Infants, Children, and Adolescents. 8th ed. Baltimore, MD: Williams & Wilkins; 2012. pp. 1147-1160

[49] Paladini D, Volpe P, Marasini M, et al. Diagnosis, characterization and outcome of congenitally corrected transposition of the great arteries in the fetus: A multicenter series of 30 cases. Ultrasound in Obstetrics & Gynecology. 2006;**27**:281-285

[50] Sharland G, Tingay R, Jones A, et al. Atrioventricular and ventriculoarterial discordance (congenitally corrected transposition of the great arteries): Echocardiographic features, associations, and outcome in 34 fetuses. Heart. 2005;**91**:1453-1458

[51] Chaoui R, Schneider MB, Kalache KD. Right aortic arch with vascular ring and aberrant left subclavian artery: Prenatal diagnosis assisted by three-dimensional power Doppler ultrasound. Ultrasound in Obstetrics & Gynecology. 2003;**22**:661-663

[52] DeVore GR, Sarti DA, Siassi B, et al. Prenatal diagnosis of cardiovascular malformations in the fetus with situs inversus viscerum during the second trimester of pregnancy. Journal of Clinical Ultrasound. 1986;**14**:454-457

[53] Bush A, Cole P, Hariri M, et al. Primary ciliary dyskinesia: Diagnosis and standards of care. The European Respiratory Journal. 1998;**12**:982-988

[54] Holzmann D, Ott PM, Felix H. Diagnostic approach to primary ciliary dyskinesia: A review. European Journal of Pediatrics. 2000;**159**:95-98

[55] Yoo SJ, Friedberg MK, Jaeggi E. Abnormal visceral and atrial situs and congenital heart disease. In: Yagel S, Gembruch U, Silverman N, editors. Fetal Cardiology: Embryology, Genetics, Physiology, Echocardiographic Evaluation, Diagnosis and Perinatal Management of Cardiac Diseases. New York, NY: Informa Healthcare; 2008. pp. 347-362

[56] Brown DW, Geva T. Anomalies of the pulmonary veins. In: Allen HD, Driscoll DJ, Shaddy RE, et al, editors. Moss and Adams' Heart Disease in Infants, Children, and Adolescents. 8th ed. Baltimore, MD: Williams & Wilkins; 2012. pp. 809-839

[57] Seale AN, Carvalho JS, Gardiner HM, et al. Total anomalous pulmonary venous connection: Impact of prenatal diagnosis. Ultrasound in Obstetrics & Gynecology. 2012;**40**:310-318