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ULTRASONOGRAPHY FOR FETAL CARDIAC ANOMALIES

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OBJECTIVE:

Fetal cardiac anomalies are common, with half of them being lethal and/or requiring complex surgeries after birth. Early detection of these anomalies enables early referral to tertiary care centers with adequate expertise. A routine antenatal ultrasound performed between18 and 22 weeks enables detection of most of these malformations. Further comprehensiveevaluation can be performed with a dedicated fetal echocardiography, particularly in high-risk pregnanciesand incases withextracardiac anomalies.

This review illustratesthe various sonographic techniques for evaluation of fetal heart and the imaging appearanceof various fetal cardiac anomalies, and prenatal counselling for the same.

Keywords:

Anomalies, cardiac, fetal, heart, malformations, ultrasound

Congenital cardiac disease is seenin2–6.5of1000livebirthsandisa major cause of morbidity andmortality,withhalfofthesecasesbeinglethalorrequiringsurgicalcorrection.Environmental,genetic,andchromosomalabnormalitiesarebelievedtobecausesofcongenitalcardiacdefects,withahigherincidenceamonginfantswithaffectedsiblingsormother.Extracardiacabnormalitiesareassociatedwith25%ofthesecases[1,2].

Detectionofcardiacanomaliescanbechallengingandistypicallydonebyfetalcardiacultrasoundperformedbetween18and22weeks.Transvaginalscancandetectanomaliesevenat12–13weeks.Detailedfetalechocardiographyisperformedinhigh-riskcases,whichcouldbearesultoffetal(extracardiac anomalies, increased nuchal translucency,hydrops,orpolyhydramnios),maternal(teratogenexposure,metabolic disorders,congenitalheartdefect,folicaciddeficiency,orautoantibodies),orfamilial(siblingorfatherwithcongenitalheartdefectandMendeliansyndromes)factors.Detectionofanomaliesalterstheobstetriccourseandoutcome,including reassurance,termination,fetaltherapy,modeofdelivery,andpostnatalreferraltoatertiarycarecenterwithadvancedexpertise inmanagementofthesepatients[2].

This article illustrates the imaging techniques and imaging appearance of variousfetalcardiacanomaliesseenwithvarioussonographic techniques, and prenatal counselling for the same.

Fetal cardiac Ultrasound:

The first step in fetal cardiac ultrasound isto evaluate the orientation of the fetus within the maternal abdomenthat is, fetal laterality (presentation and lie). Orientation isassessed from a transverse section of the fetal abdomen. If the fetal head is found below this level and the spine is posterior, thenthe left side of the fetus should be located onthe right side of the maternal abdomen andthe stomach should be on this side (Fig. 1A).On the other hand, if the fetal head is foundabove this level and the spine is posterior,then the right side of the fetus is located onthe right side of the maternal abdomen andthestomachislocatedontheoppositeside[3](Fig.1B).Situsisthepositionthatanorganoccupiesinrelationtothebilateralbodysymmetry.Foratrial situs, this is the relationship of the atriawithrespecttoeachother.Thisisconventionallyestablishedbynotingthearrange

mentofthe aorta and inferior vena cava (IVC) at thelevel of the diaphragm, because great vesselarrangementissimilartothoracicandatrial situs. In situssolitus, the IVC is anterior andto the right of the aorta (Fig. 2A). In situs inversus, there is a mirror image pattern, withthe aorta to the right of the IVC (Fig. 2B). Insitus ambiguous, the aorta and IVC are located on the same side of the spine in rightisomerism (Fig.2C)andtheaortaiscentrallylocated,withaninterruptedIVCinleftisomerism[3] (Fig.2D).

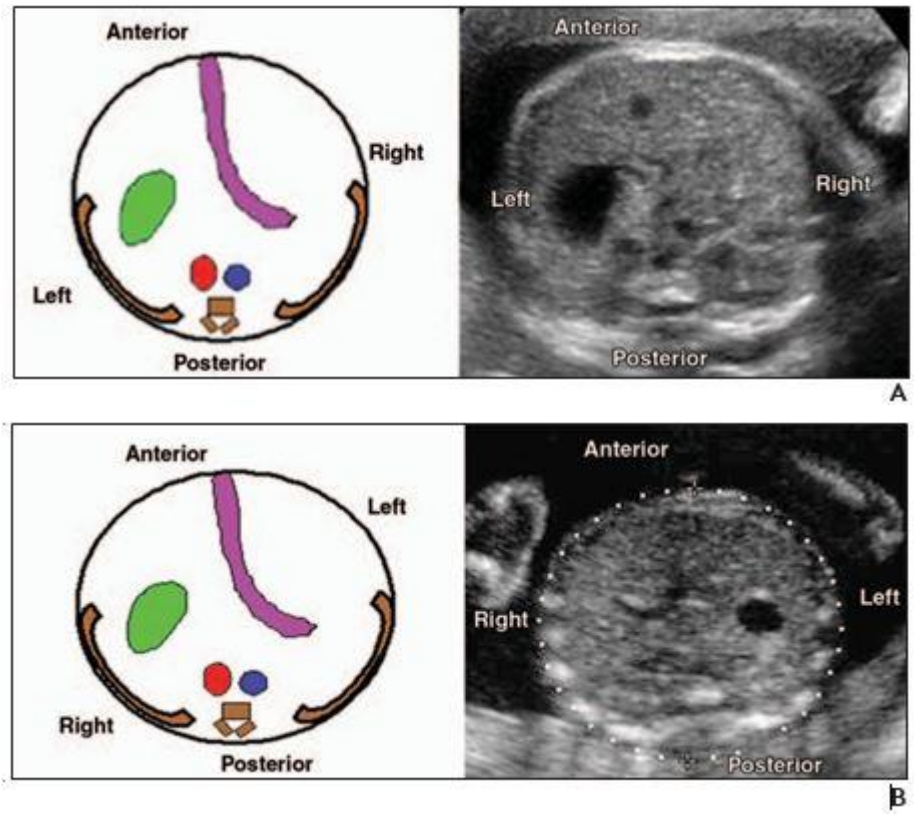


Fig.1—Establishinglaterality.Schematicdiagramand ultrasound through abdomen show spine andstomach.
A,Inthisfetus,fetalheadwaslocatedbelowplaneof abdomen (vertex presentation), making left sidelower; hence, stomach was located on left side offetus.
B,Inthisfetus,fetalheadwaslocatedaboveplaneofabdominalimage,makingleftsideupper;hence,stomachwaslocatedonrightsideoffetus.

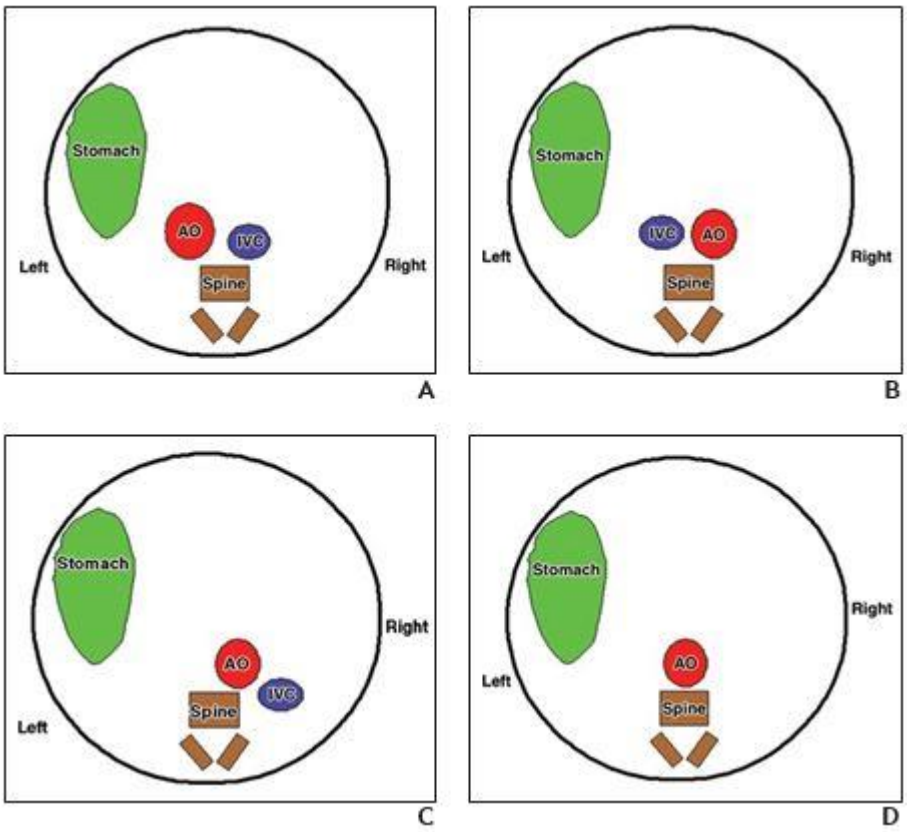
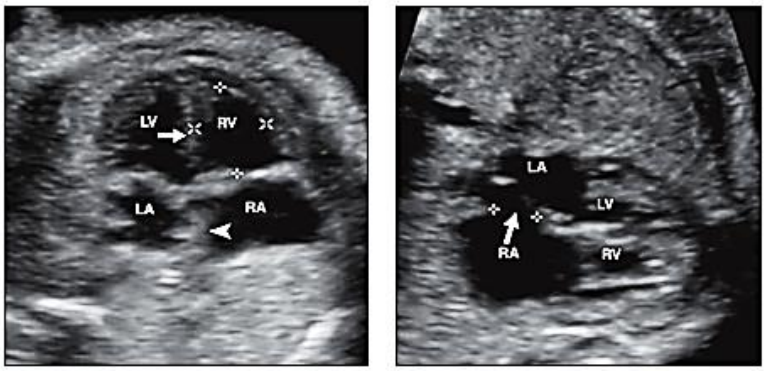


Fig. 2—Establishing situs and atrial arrangement.Diagrams represent axial images acquired at level oflowerthoracicspine.
A,Insitussolitus,aorta(AO)islocatedtoleftofinferiorvenacava(IVC).
B,Insitusinversus,AOislocatedtorightofIVC.
C,Inrightisomerism,AOandIVCarelocatedonsamesideofspine.
D,Inleftisomerism,AOislocatedcentrally,andIVCisnotvisualizedbecauseitisinterrupted.

Views in Fetal Cardiac Ultrasound:

The basic view performed in cardiac ultrasoundisthefour-chamberview[4],whichcan detect 43–96% of fetal anomalies [1].“Extended basic views” of the left ventricularoutflowtract(LVOT)andrightventricular outflow tract (RVOT) increase thesensitivityforthedetectionofanomalies.The four-chamber view is obtained by atransverse projection through the fetal thoraxabove the level of the diaphragm, either apical(paralleltotheinterventricularseptum)or subcostal (perpendicular to the interventricular septum). This view shows the twoatria and ventricles along with atrioventricular (AV) valves (mitral and tricuspid) andsepta (interventricular and interatrial) (Fig.3). The cardiac position and axis are determined in this projection. In levocardia, theheart is located within the left chest, with theapexpointingtotheleft;indextrocardia,itislocated within the right chest with the apexpointingtotheright;andinmesocardia,itis centrally located

with the apex pointinganteriorly. The cardiac axis is calculated from alinedrawnfromtheposteriorspinetotheanterior sternum (spinosternal line). The ventricularseptumtypicallyintersectsthislineat 40–45°. Cardiac axis may be altered in intracardiacconditions(Ebstein'sanomalyand tetralogyofFallot)orextracardiacconditions causing mass effect or as a result of accompanyingpulmonaryhypoplasia[5].



A B

Fig. 3—Standard cardiac view.
A, Basic four-chamber view, which shows right ventricle (RV), left ventricle (LV), right atrium (RA), and left atrium (LA). Interventricular septum (*arrow*) is also seen in this view that is acquired perpendicular to ultrasound beam. Interatrial septum (*arrowhead*) is visualized.
B, Four-chamber view in another fetus shows patent foramen ovale (*arrow*).

Cardiacanatomyistypically evaluated using a sequential segmental approach,whichdependsonmorphologicidentification of the atria, ventricles, and great arteries, not on their spatial relationship [3]. Themorphologic right atrium (RA) has a triangular appendage, whereas the morphologicleft atrium (LA) has a hook-shaped appendage.Differencesbetweenthemorphologic right ventricle (RV) and the left ventricle(LV)arelistedinTable1[5].Thetricuspid valve opens into the RV and the mitral valveopens into the LV, with the septal leaflet oftricuspid valve inserting more apically thanthemitralvalve.Occasionally,valvesmaybeimperforate, common, straddling, or overriding. The AV junction is the continuation ofatriawiththeventricularchamber.Inthetypical biventricular connection, each atriumconnects with a ventricular chamber, whichcould either be concordant (i.e., RA-RV andLA- LV), discordant (i.e., RA-LV and LA-RV), or ambiguous (i.e., isomeric RA or LAwithRVtotherightorleftofLV).Inuniventricular connection, one or two atrial chambers connect with a single ventricular cavity[3]. There are two arterial valves, the aorticand pulmonary, which connect the LV to theaorta and the RV to the pulmonary artery(PA),respectively.Ventriculoarterialconnections can be concordant (i.e., aorta fromLVandPAfromRV),discordant(i.e.,aorta from RV and PA from LV), double outlet(i.e.,>50%ofeachgreatarteryconnectedto same ventricular cavity), and single outlet (i.e., only one arterial trunk connected tothe ventricle).

TheLVOTviewisobtainedbya45°tiltofthe transducer from the four-chamber viewperpendicular to the septum, to an obliqueplanefromthefetalupperleftquadrantoftheabdomentothefetalrightshoulder.Theaortaoriginates from the LVOT (Fig. 4A) and dis-

tallygivesoffthegreatvesselsoftheheadandneck. Membranous septum is also visualizedinthisview.TheRVOTviewcanbeobtainedby further rotation in the same direction andgentlerockingofthetransducerfromtheLVOTview.ThePAisseenexitingfromtheRV (Fig. 4B), dividing into the right PA andleftPA(Fig.4C)andcontinuingastheductusarteriosus, which opens into the descendingthoracicaorta(Fig.5).Theascendingaortaisseencentrally,wrappedbytheRVandPA. In both of these projections, a normal aortaand PA are perpendicular to each other. Furtherrightwardrotationresultsinshort-axisviewsof the ventricles and thorax. Further rotationtoward the left fetal shoulder shows the aortaasacentralcircledrapedbythePAanteriorlyandtotheleft.Rotationfromtheleftshoulderto the right hemithorax shows the aortic archand its branches (Fig. 5A) and ductus arteriosus (Fig. 5B). The ductal arch (RVOT, PA,and ductus arteriosus) is broader and flatterthantheaorticarch.

Acomprehensivesetoffiveshort-axisprojections can also be acquired, with theventricular septum parallel to the ultrasoundbeam. These views are better at detection ofconotruncal abnormalities that are missed bythe routine views. From caudad to cephalad,these views are the abdomen at the level ofthe stomach to identify situs; the four-chamber view; the five-chamber view, which includes a centrally placed aorta; PA bifurcation, with the PA originating from the RVlocated on the left side and crossing the aorta to lie to the left side and anterior of aorta;andthethree-vesselview,whichincludesthemain PA, ascending aorta, and superior venacava (SVC) from the left anterior to the rightposterior aspect of the thorax [5] (Fig. 6).The SVC typically opens into the RA (Fig.5C). PulmonaryveinstypicallydrainintotheLA,but anomalous veins can drain into systemicveins,eitherpartiallyortotally[3](Fig.5D)

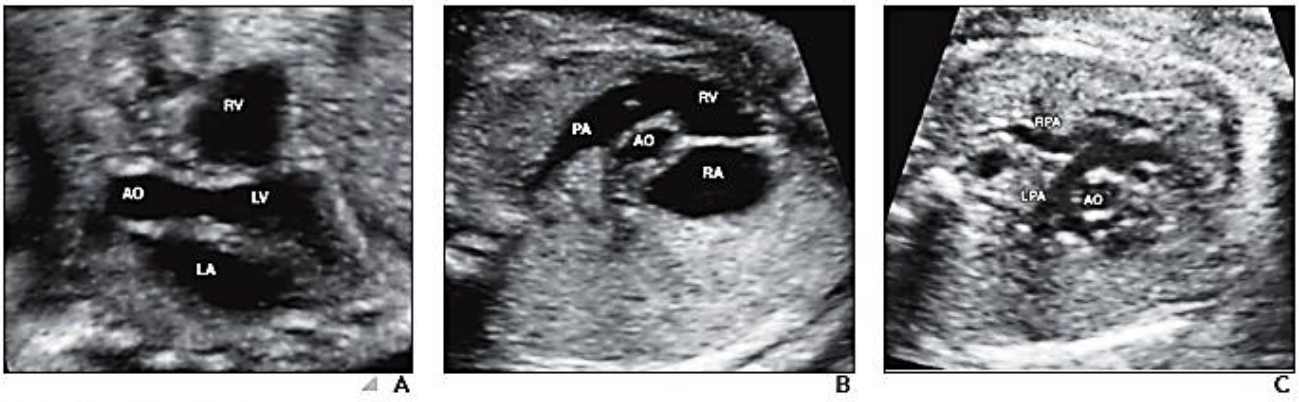


Fig. 4—Extended cardiac views.
A. left ventricular outflow tract view shows left ventricle (LV), which gives origin to ascending aorta (AO). Right ventricular (RV) outflow tract and left atrium (LA) are also seen.
B. Right ventricular outflow tract view shows RV, giving origin to main pulmonary artery (PA). AO is seen as circular structure and is perpendicular to PA.
C. Further superiorly, right pulmonary artery (RPA) and left pulmonary artery (LPA) are seen.

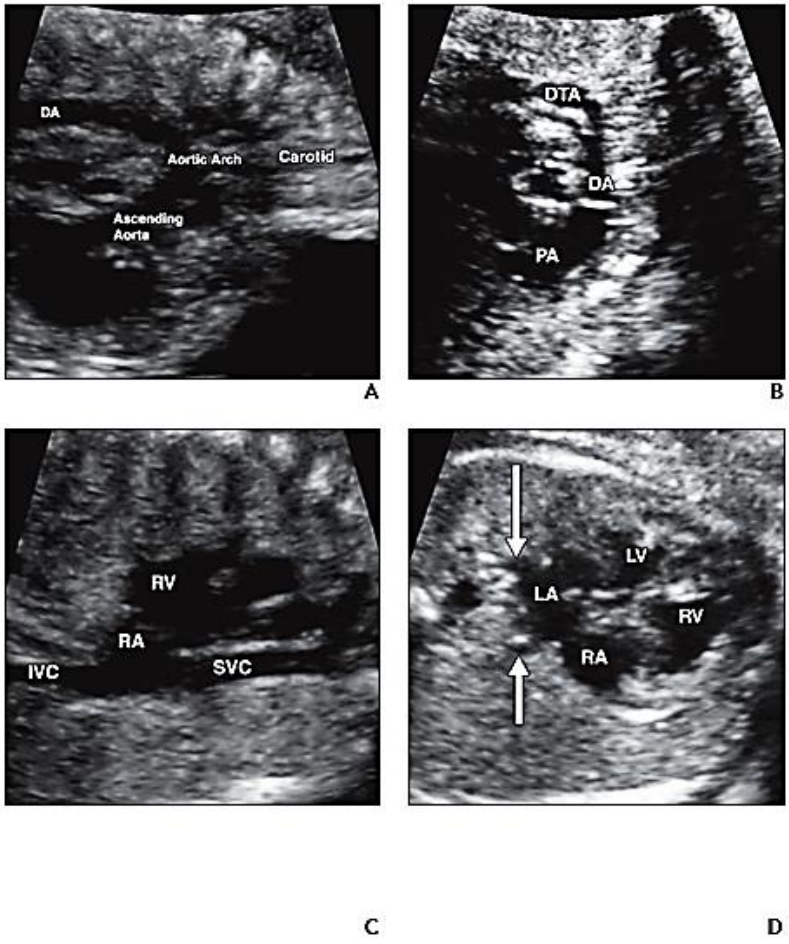


Fig. 5—Vascular structures.
A, Aortic arch can be seen giving off branches to head and neck and continuing as descending thoracic aorta (DTA). DA = ductus arteriosus.
B, DA connects pulmonary artery (PA) to DTA.
C, Superior vena cava (SVC) and inferior vena cava (IVC) open into morphologic right atrium (RA). RV = right ventricle.
D, Pulmonary veins (*arrows*) open into morphologic left atrium (LA). LV = left ventricle.

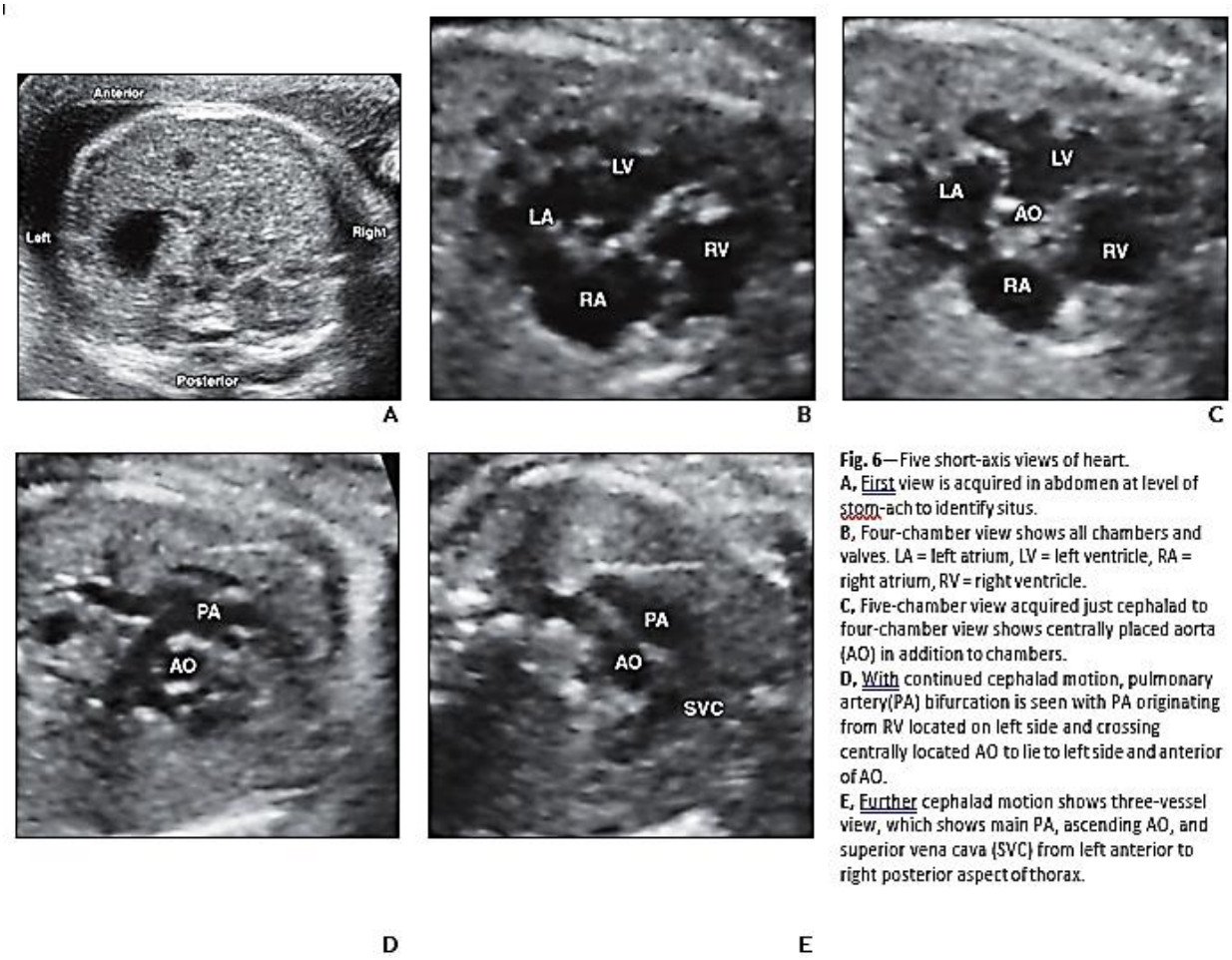


Fig. 6—Five short-axis views of heart.
A, First view is acquired in abdomen at level of stom-ach to identify situs.
B, Four-chamber view shows all chambers and valves. LA = left atrium, LV = left ventricle, RA = right atrium, RV = right ventricle.
C, Five-chamber view acquired just cephalad to four-chamber view shows centrally placed aorta (AO) in addition to chambers.
D, With continued cephalad motion, pulmonary artery(PA) bifurcation is seen with PA originating from RV located on left side and crossing centrally located AO to lie to left side and anterior of AO.
E, Further cephalad motion shows three-vessel view, which shows main PA, ascending AO, and superior vena cava (SVC) from left anterior to right posterior aspect of thorax.

Measurements:

The cardiac chambers and vascular structures are measured and can be compared with normalized charts. The RV and LV typically are of the same size, with a 1:1 ratio. The cardio thoracic ratio is the ratio between the cardiac and thoracic circumferences, which normally measures 0.5 [6]. Normalized charts at various gestational age sexist for the RV: LV ratio, LV wall thickness, septal wall thickness, left atrial dimension, PA diameter, and aortic root diameter. A small heart is seen as a result of hypoplasia (LV or RV or both) or of compression fromex trinsic masses, whereas a large heart can be seen in various congenital abnormalities, pericardial effusion, aneurysms, cardio myopathies, and ortumors. The PA diameter is typically larger than the aorta by approximately 10%.However,asa general rule, the two can be considered to be similar in size and any discrepancy in size should be concerning.

M-mode Ultrasound:

M-mode ultrasound is a 2D image of motion over time that is used for evaluation offetalheartmotion,heartrate,wallthickness, chamber size, and motion of the valvesor myocardium. Fetal heart rate and rhythmcan be evaluated using M-mode ultrasoundthroughtheatrialandventricularwall,aboveandbelowtheAVvalve,respectively.Chamber size and function are evaluated by focusingatthelevelofAVvalves[6].AVconcordance can be evaluated by using M-modeultrasound through both atrium and ventricle at the same time. Normal fetal heartbeatsare175beats/minat8weeks,140beats/minat 20 weeks, and 130 beats/min at term [5],witharegular 1:1AVrhythm [5].

Colour-Flow Pulsed Doppler and Doppler Tissue Imaging:

Color Doppler can be used to detect vascular flow through cardiac chambers, vascular structures, and septal defects. It alsosignificantly reduces the time required forDoppler examination of the heart, becauseinterrogation of vascular structures becomeseasier [1]. The direction of the flow can beestablished, which is useful in the detectionandquantificationofregurgitationandstenosis. Reversal of the flow through a valve indicates regurgitation. The presence of aliasingincolorDopplerindicateshighvelocitiessuggestive of stenosis. If pulsed Doppler isappliedatthispoint,highvelocitywithspectralbroadeningcanbeseen.Normalpeak velocitythroughtheAVvalvesis30–60cm/s throughout gestation, and that throughthearterialvalvesis25cm/sat12weeksand60–100 cm/s by term [6]. Color Doppler isalso used in the evaluation of pulmonary andsystemic venous connections and small septaldefects.ColorDopplertissueimaginghasbeen used recently for evaluating high-amplitude low-velocity signals, such as withinthemovingmyocardium.Thiscanbeusedto encode the direction of myocardial motion, which is particularly useful in the assessment ofarrhythmias[7].

Three and Four-Dimensional Ultrasound:

In 3D ultrasound, volumetric data are acquired from a single window using few secondsofscanning,whicharesubsequentlyused for reconstruction of multiple views inany plane. This reduces the overall scanningtimeandthe

operatorandwindowdependence,inaddition to improving the assessment of cardiac anatomy. Planes that are not accessiblein 2D scanning, such as the interventricularseptum or coronal plane, can be reconstructed. Volume-rendered or surface-shaded images give an illusion of depth, which mightbeusefulinthedetectionofcomplexanomalies, such as those involving the conotruncalseptum.Alternatively,theimagescanbedisplayedasthreeorfoursimultaneousmultiplanarreformattedprojectionswiththeabilitytomovethroughthevolumes.Three-dimensional quantitative measurements are more accurateandreproduciblethan2Dtechniques.Acquisitionoftemporalinformationwithcardiacgatingenablesdisplayoftheseimagesascineloopsinmultipleplanes(4Dimaging),whichis useful in the evaluation of cardiac motion,cardiac function, valvular function, volumes,and cardiac output. Three-dimensional imaging of regurgitation and stenotic jets is possible when 3D ultrasound is combined withcolororpowerDopplerimaging.Three-dimensional color-flow angiography can revealcomplexcardiovascularanatomy,anomalousvessels,andsmallseptaldefects [7].

Atrial Septal Defect:

Atrialseptaldefect(ASD)ischaracterizedby defect in a portion of the atrial septum. Itis the fifth most common congenital heartdisease, seen in 1 of 1500 live births [1], andis caused by abnormal tissue resorption anddeposition during development of the atrialseptum.Accordingtoitslocation,itisclassified as ostiumsecundum (midatrial septum),ostiumprimum(loweratrialseptum)(Fig.7),sinusvenosus(outsidetheatrialseptumin the wall separating the SVC or IVC fromthe LA), and coronary sinus defect, whichcan be partial or complete. ASD may be difficult to visualize in a fetus because of thepresence of foramen ovale. However, withhigh-resolution ultrasound, the septum primum is seen in the four-chamber view as acircular or linear structure with a loose pocketconfiguration,andtheseptumsecundumisseenasathickstationarystructure withthe foramen ovale opening into it. Normalforamen ovale measures almost same as theaorticroot,withthedifferencebeing1mmorless[1](Fig.3B).Theforaminalflapof theforamen ovale is seen moving into the LA attwice the heart rate. A secundum defect isseen as a larger defect in the central portionof the atrial septum or a deficient foramenflap. A primum defect is seen in the lowerpartoftheatrialseptum.

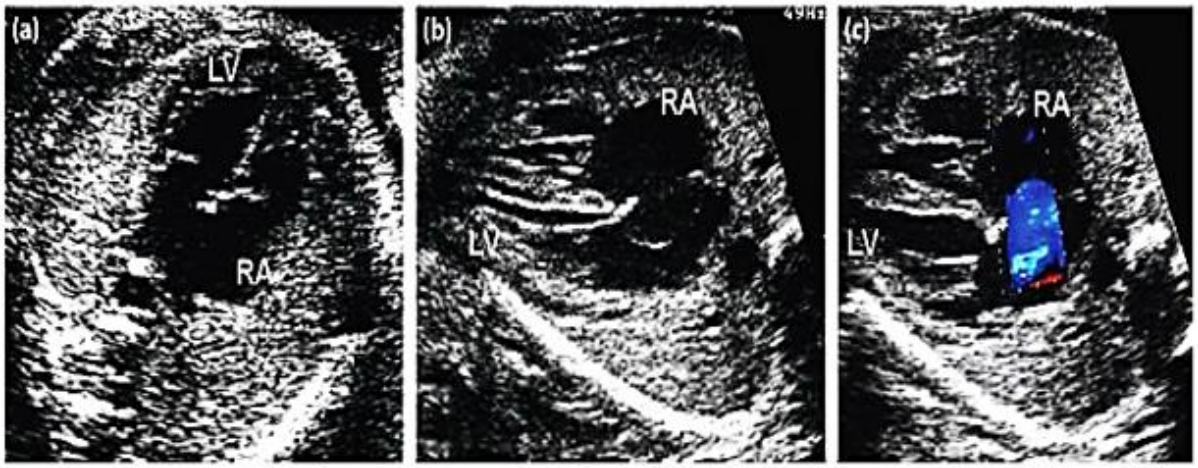


Fig. 7. Atrial Septal Defect: two cases of ASD confirmed at birth.

a. On the apical four-chamber view, an extremely wide (8 mm) foramen ovale can be seen.
b. Also, in this case, on the transverse four chamber view, the large size of the foramen ovale is evident; the flap is seen in the left atrium.
c. Same case as in (b): colour Doppler demonstrates the extent of the shunt.
LV: left ventricle RA: right atrium

Counseling:

If ASD is isolated, as in most instances, survival and quality of life are unaffected, regardless of the need for an interventional procedure. Only in the unlikely case in which an ASD is detected late, when it has already caused irreversible pulmonary hypertension, does the life expectancy decrease and there may be severe complications adversely affecting survival and quality of life.

Ventricular Septal defect:

Ventricularseptaldefect(VSD)isthemostcommoncongenitalheartdisease,seenin1.5–3.5 per 1000 live births, and accounting for30% of all cardiac anomalies [8]. The defectismostcommonly(80%)seeninthemembranous septum and less commonly in the muscular,outlet,orinletportions.Defectscanbevariableinsize.VSDisbestseeninafour-chamberviewasdiscontinuityintheventricularseptum,particularlytheinletdefects. Theventricularseptumisideallyevaluatedinimages acquired perpendicular to the interventricular septumbecause a pseudo-VSD, as a result of signal drop-out, can beseen in the superior aspect of images parallel to the ultrasound beam [8]. Membranousseptum is also seen in the LVOT view. Out-let defects are best seen when the transducer is angled anteriorly [3]. Small defects canbe difficult to detect, particularly in the perimembranous portion, but Doppler imagingcan show flow across the defect. In isolatedVSD,bidirectionalshuntingwithright-to-

leftshuntduringsystoleandleft-to-rightshuntingindiastoleisseen,butinVSDassociatedwithotheranomalies,unidirectionalshuntingmaybeseen.Smalldefectsmayclose,butlargedefectsrequiresurgicalclosure.

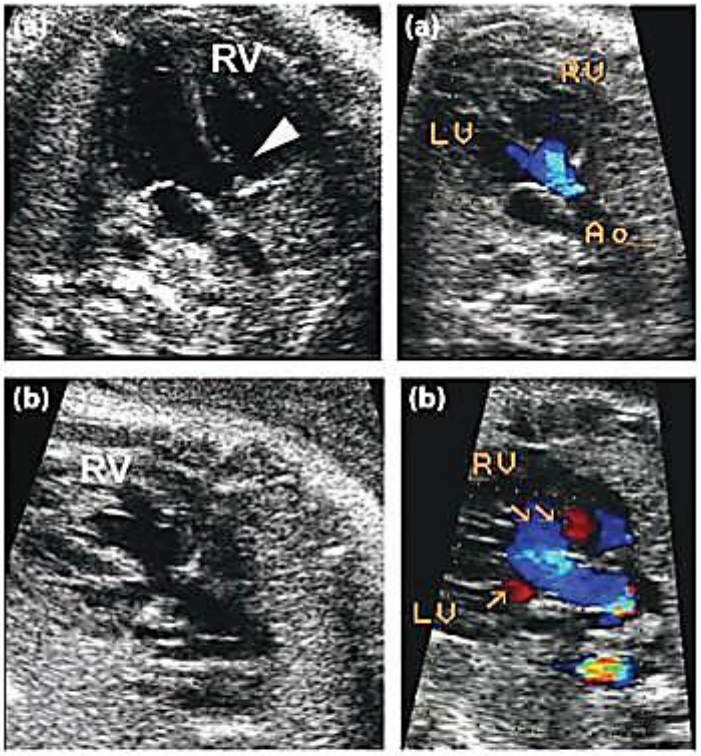


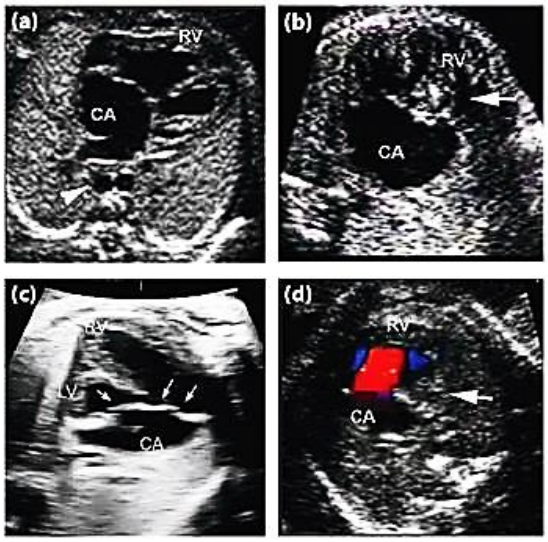
Figure . Ventricular septal defect (VSD). VSDs not detectable on the four-chamber view but only on the left outflow tract view which include (a) outlet subaortic perimembranous VSD (color Doppler demonstration on the right, 31 weeks of gestation); and (b) malalignment VSD, with the ascending aorta overriding the VSD (color Doppler demonstration on the right, 27 weeks of gestation). Ao: aorta; LV: left ventricle; RV: right ventricle.

Counseling:

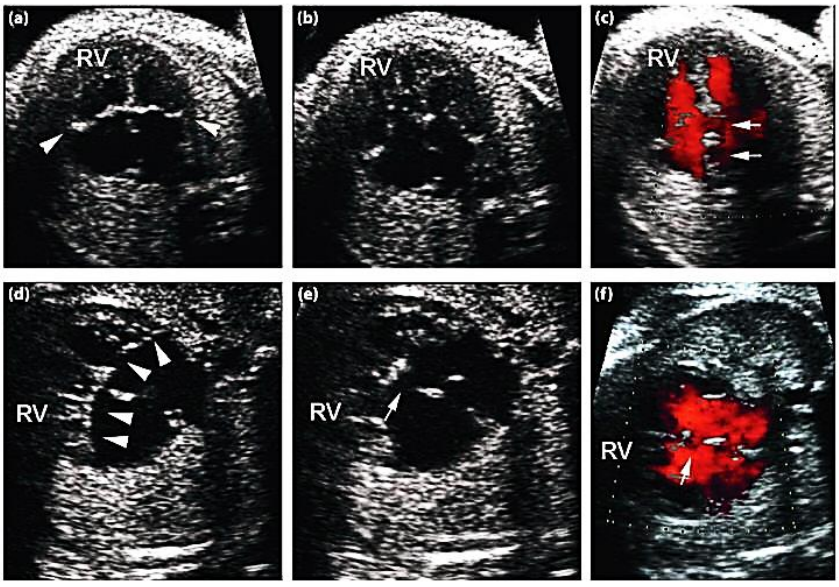
In postnatal series, the prognosis for isolated VSDs is extremely favourable, with a high rate of spontaneous closure (upto 90% of cases, if the smallest muscular defects are included) and normal life expectancy both for untreated nonrestrictive VSDs and for large ones, if operated. In contrast, prenatal series show lower survival rates, as is often the case when comparing prenatal and postnatal data for CHD: the intrauterine mortality rate can reach 11% and the neonatal mortality rate 31%, with a five year survival rate of 40%.

AV Septal Defect:

AV septal defect (AV canal defect or endocardialcushiondefect)iscausedbyfailureof fusion of the endocardial cushion, resultingindefectsoftheatrialostiumprimum, theventricularinletseptumcommonAVvalve, and the biventricular AV connections.AV septal defect accounts for 2–7% of con-genital heart defects and is seen in 0.19–0.56per 1000 live births [8]. It is associated withtrisomy 21 syndrome, left atrial isomerism,hypoplasticleftheart,pulmonarystenosis,coarctation, tetralogy, complete heart block,and extracardiac anomalies. There are twotypes:thecompletetype(97%ofcases),withcommon valvular orifice, and the incompletetype, with separate right and left valve orifices. The valve of common AV junction hasfive leaflets, which are separate in the completetype,buttwoleafletsareconnectedbynarrowtissueintheincompletetype.It is associated with a cleft in the anterior mitral leaflet. Free regurgitation is seen acrossthe common AV valve [8]. Direct shuntingmay be seen from the LV into the RA. In severe forms, all four chambers communicate,causing left-to-right and right-to-left shunt.Ultrasound shows a defect in the endocardial cushion, with an inlet VSD and primumASD (Fig.) associated with a single abnormal AV valve that has a T-shaped arrangement.ColorDopplershowsopenflowacrossthedefectandabnormalAV valve.



Atrioventricular septal defect (AVSD) complete, unbalanced in atrial isomerism (21–22 weeks of gestation). (a) Moderate ventricular disproportion (right ventricle larger than left). However, the azygos continuation (arrowhead) adjacent to the descending thoracic aorta should arouse suspicion of a situs abnormality. (b) Unbalanced AVSD, with severe hypoplasia of the right ventricle (arrow). (c) Unbalanced AVSD, with severe hypoplasia of the left ventricle (arrow). (d) In the same case as in (c), color Doppler demonstrates overriding of the atrioventricular valve and obstruction of the left part of the valve, responsible for the hypoplasia. CA: common atrium; RV: right ventricle.

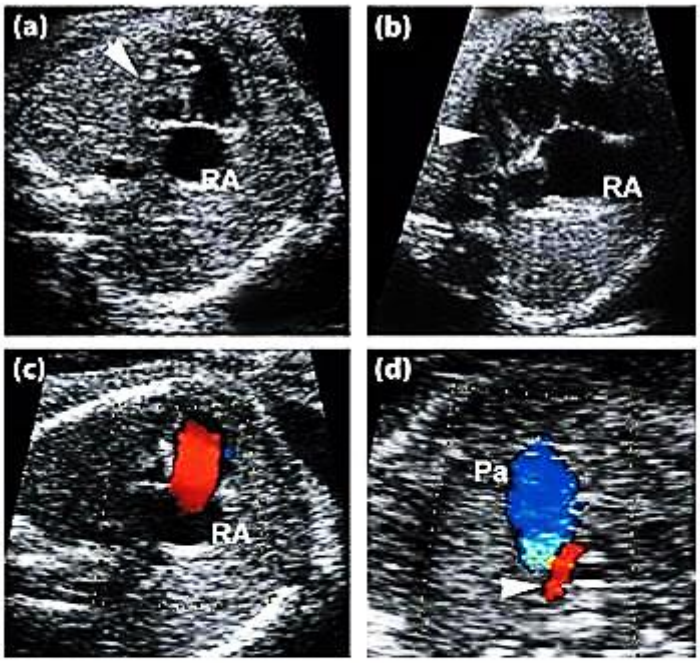


Atrioventricular septal defect (AVSD) partial (29 weeks of gestation). This is characterized by an atrial septal defect (ASD) of the ostium primum type (absence of the septum primum) with two atrioventricular valves, which do not show the normal offset aspect. (a,b) On 2D ultrasound, during systole (a), on the apical four-chamber view, the lack of the normal offset aspect is seen (arrowheads). In diastole (b), two separate atrioventricular valves are demonstrated. (c) Color Doppler helps to confirm the presence of the septum primum defect and, when present, of the VSD (arrows). (d) In systole, but on the transverse four-chamber view, the loss of the offset appearance of the atrioventricular valves is confirmed (arrowheads). (e) In diastole, the ostium primum defect is shown, just above the atrioventricular plane (arrow). (f) Color Doppler demonstrates flow across the ostium primum defect (arrow), across the normal foramen ovale (to the right of the ostium primum), and across the small VSD (to the left of the ostium primum). RV: right ventricle.

Hypoplastic Left Heart Syndrome:

Hypoplastic left heart syndrome is characterizedbyhypoplasticleft-sidedcardiacstructures, including the LV, mitral valve,aortic valve, and aorta. It accounts for 2–4%ofcongenitalcardiacdefectsandisseenin 0.16–0.25 per

1000 live births [8]. It ismore common in boys and is caused by de-creased flow in and out of the LV during development (e.g., mitral or aortic stenosis oratresia). Blood flow to the systemic circulation (coronary arteries, brain, liver, and kidneys) in these patients is dependent on flowthrough the ductus arteriosus. It is associatedwith aortic coarctation in 80% of cases [10].Onultrasound, the LV issmall (LV:RV ratio< 1) in size (Fig.); the ventricular septummakes an angle of 90° with the spinosternalline, and the aortic outflow is smaller thanthe pulmonary outflow tract. Mitral and aortic valves are hypoplastic or atretic. A singlearea of flow is seen at the AV level and bidirectional flow at the proximal aorta becauseof distal aorticcoarctation [1].



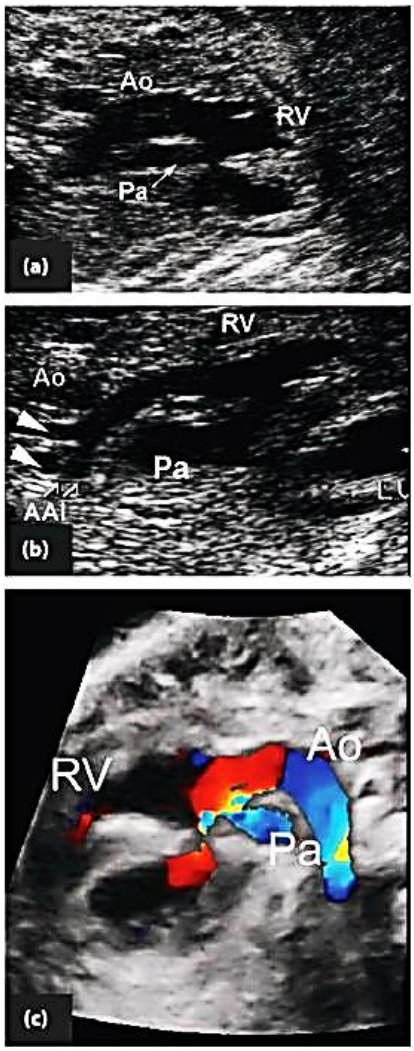
Hypoplastic left heart syndrome. (a) At 22 weeks of gestation, on the four-chamber view, the severe hypoplasia of the left ventricle, transformed into a virtual cavity due to the mitral atresia (arrowhead), is evident. (b) At 32 weeks of gestation, there is severe hypoplasia of the left ventricle, which is slitlike (arrowhead). (c) In the same case as in (a), color Doppler demonstrates the mitral atresia (absence of left ventricular filling). (d) On the three-vessel view, the reverse flow across the hypoplastic aortic arch (in red, arrowhead) (associated tubular hypoplasia) confirms the aortic atresia.

Counseling:

Despite the recent brilliant achievements of paediatric cardio surgery, HLHS continues to be congenital cardiac defect with highest mortality.

Double Outlet Right Ventricle:

DoubleoutletRV(DORV)ischaracterizedbytheoriginofmorethan50%ofboththeaorta and PA from the RV and is caused by abnormalspiralingofthetruncusarteriosusandthearrestofmembranousseptalformation.Itaccounts for less than 1% of congenital heartdefectsandisseenin0.08–0.16per1000livebirths[8].TherearefourtypesofDORV:aortaparalleltothePAandtoitsright(64%),whichresembles tetralogy of Fallot; aorta anteriorand to the right of the PA (26%), resemblingD-transposition of great arteries; aorta anteriorandtotheleftoftheaorta(7%),resemblingL-transposition of great arteries; and aorta posteriorandtotherightofthePA(3%).DORVisalmostalwaysassociatedwithVSD, whichprovidestheonlyoutletfromtheLV.Itisassociatedwithmaternaldiabetesoralcoholintakeandothercardiacdefects,suchasLVhypoplasia,mitralvalvestenosisoratresia,aorticvalvestenosis,aorticcoarctationorinterruption,andcoronaryarteryanomalies.DORVisbestseenin short-axis views, where the aorta and pulmonaryarteriesdonotcrossandboththevesselsarisefromtheRVandareparalleltoeachother(Fig.).Demonstrationoftheoriginofboththevesselsfromthesamesideofventricularseptumisessentialtodifferentiate DORVfromtranspositionofgreatarteries[8].



Double-outlet right ventricle (DORV). This comprises a spectrum of anomalies of the great vessels, which may have different sizes due to left or right outflow obstruction, in different spatial relationships (see text of this chapter). (a) DORV with an anterior malposed aorta and a posterior moderately stenotic pulmonary artery (arrow); this is one of the most common arrangements seen in the fetus. (b) DORV with an anterior malposed aorta, which is also reduced in size due to a concurrent interruption of the aortic arch; note the straighter course of the aorta and the "V" shape of the first neck vessels (arrowheads). (c) Color Doppler can be used to confirm the connection of the great vessels with the anterior ventricle, if there is any remaining doubt regarding the differential diagnosis with transposition of the great arteries. AAI: aortic arch interruption; Ao: ascending aorta; Pa: main pulmonary artery; RV: right ventricle.

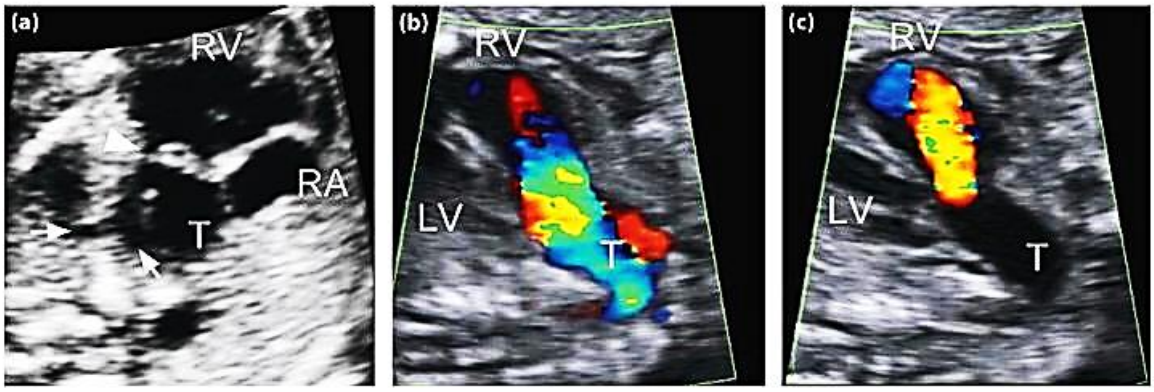
Counseling:

The overall survival of DORV detected prenatally is 46-50% (if terminations of pregnancy are excluded), due to the strong association with aneuploidy and extra cardiac anomalies.

Truncus Arteriosus:

Truncus arteriosus is characterized by asingle arterial trunk that feeds

the systemicpulmonary circulation and coronary arterieswith a single semilunar valve. It accounts for1–2%ofcongenitalcardiacdefects,isseenin0.08–0.16per1000livebirths[8],andis causedbyfailureoffusionanddescentoftheconotruncal ridge. It almost always straddlesaVSDandreceivesbloodfromboththeventricles but rarely originates almost completely from the RV or LV. There are four types(CollettEdwardsclassification)basedonthe level of origin of the aorta and pulmonary arteries [9]. An admixture of oxygenated and deoxygenated blood in the commontrunk results in subnormal systemic oxygenation. The ductus arteriosus is not necessaryfor systemic flow and therefore does not fully develop. On ultrasound, a single arterialtrunk is seen overriding the interventricularseptum, with an associated VSD, and thereareseveralbranchesconnectingwiththeaortaandpulmonaryvasculature(Fig.)



Common arterial trunk (CAT), type I, 23 weeks of gestation. (a) The main pulmonary artery with the two hypoplastic branches (arrowheads) is seen branching off the truncus (T) just above the truncal valve (arrowhead). (b) On the left ouflow tract view, color Doppler demonstrates (during systole) moderate stenosis of the truncal valve (aliasing and turbulence), due to a highly dysplastic truncal valve. Straddling of the truncus toward the right ventricle is also visible. (c) During diastole, massive insufficiency of the dysplastic truncal valve is visible. Note how the truncus is prevalently connected to the right ventricle due to the straddling. LV: left ventricle; RV: right ventricle; T: arterial trunk.

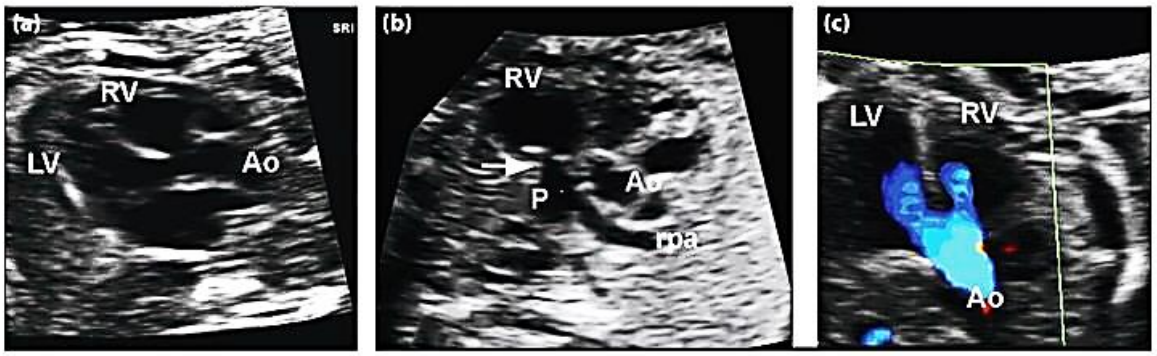
Counseling:

Prognosis depends on the presence of extracardiac and chromosomal anomalies and of unfavourable cardiac anatomy. (e.g. severe truncal valve regurgitation, IAA, and straddling with ventricular hypoplasia)

Tetralogy of Fallot:

TetralogyofFallotischaracterizedbynarrowing of the RVOT, VSD, overriding aorta,andrightventricularhypertrophy.Itaccountsfor 5–10% of congenital cardiac defects andis seen in 0.24–0.56 per 1000 live births [8].It is caused by anterior displacement of theconotruncus, resulting in unequal division ofconus into a small anterior RV portion andlargeposteriorLVportion.Theincompleteclosureoftheseptumresultsinaortic overriding.It is associated with chromosomal and extra-

cardiacabnormalities.Onultrasound,theaortaisseenstraddlingalargemembranousVSD(Fig. 9). Depending on the size of the PA, itmay not be easily seen and the normal crossingofaortaandpulmonaryarteriesisnotseen.The aorta may be dilated, and the pulmonaryvalveisstenosedoratreticwithadilatedPA.BecauseofthepresenceofnormalfetalshuntsRVhypertrophyisnotseeninthefetus.



Tetralogy of Fallot (TOF): the typical signs of TOF are shown. (a) The left outflow tract view shows a malalignment VSD with an overriding aorta. (b) The right outflow tract view shows the narrowing of the pulmonary trunk, consistent with an infundibular stenosis (arrow). (c) On the left outflow tract, color Doppler demonstrates the overriding aorta draining from both ventricles. Ao: aorta; LV: left ventricle; P: pulmonary artery; rpa: right pulmonary artery; RV: right ventricle.

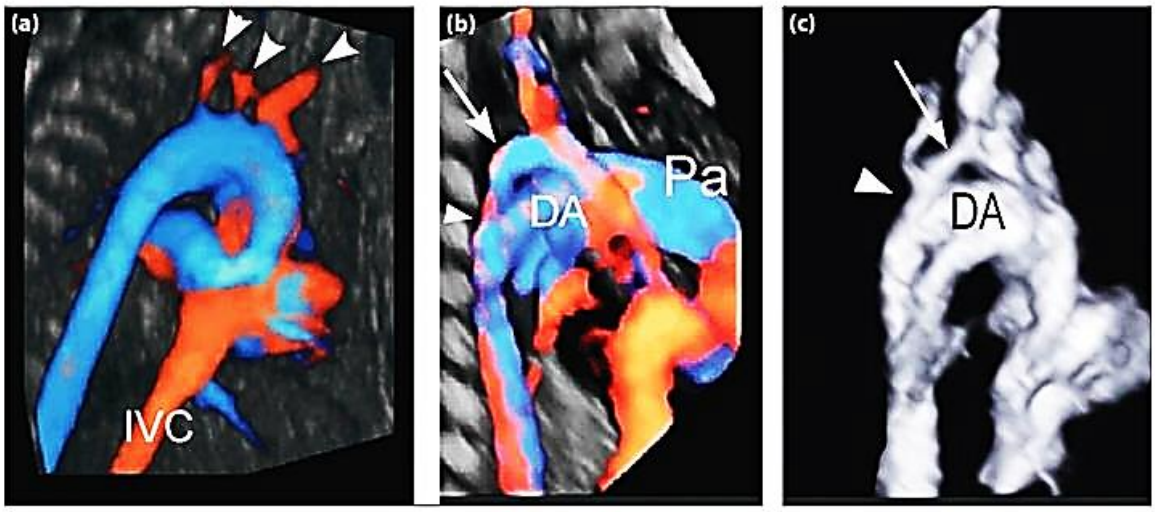
Counseling:

The overall prognosis will depend on several factors, including karyotype, associated extracardiac malformations and cardiac anatomy. Prenatal counseling should therefore take into considerations all the aspects in addition to the well-established fact that the pulmonary outflow obstruction may evolve during pregnancy. With regard to survival, case series of patients with isolated TOF report long term survival rates as high as 80-90%. In conclusion, if no unfavourable prognostic factors are found in utero and, above all, after birth,TOF is an easily correctable heart defect with excellent survival and good quality of life.

Coarctation of Aorta:

Coarctation is discrete narrowing of theaortic arch, most commonly distal to the leftsubclavian artery. It accounts for 7% of allcongenitalcardiacdefects[11]and6%ofcardiacanomaliesseenprenatally[12].It canbe associated with chromosomal abnormalities, maternal diabetes, bicuspid valve, aorticstenosis, Turner syndrome, intracranial aneurysms, VSD, ASD, Shone complex, transposition, Taussig-Bing anomaly, and aortichypoplasia. Coarctation may be difficult to visualize in ultrasound and is diagnosed whenthedistalarchissmallerthannormalizedvalues. In hypoplasia, the entire arch is small(Fig.). In addition, the ascending aorta

issmall(ratioofthePAtotheascendingaorta,>2SDabovenormal).TheLVcanbesmall whenitispartofthehypoplasticLVsyndrome.Coarctationmayprogressinutero[1].



Coarctation of the aorta. Four-dimensional echocardiography (STIC) contributes to the characterization of aortic coarctation. Volumes should be acquired with a longitudinal ventral approach for adequate imaging. (a) Normal fetus at 22 weeks of gestation. Glassbody rendering shows the normal size of the aortic arch and the three neck vessels (arrowheads). The isthmic tract shows no indentation or reduction in caliber. (b) In a fetus with aortic coarctation (26 weeks of gestation), the same view demonstrates the tapered, hypoplastic aortic arch (arrow) and the much greater size of the ductal arch. The arrowhead indicates the shelf. (c) In another case of severe aortic coarctation, B-flow demonstrates (even more clearly than with glassbody) both the tapering (arrow) and the shelf (arrowhead), which represent classic findings in severe aortic coarctation. DA: ductal arch; IVC: inferior vena cava; Pa: pulmonary artery.

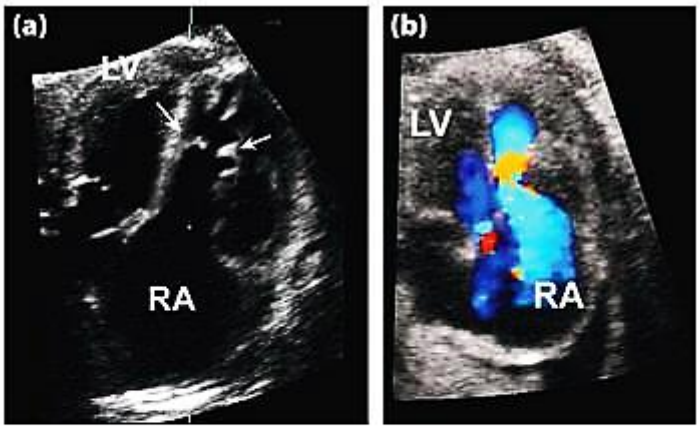
Coarctation of the aorta. (a) On the four-chamber view, the ventricular disproportion, which is an indirect sign of coarctation, is evident (the right ventricle is larger than the left). (b) The three-vessel view demonstrates the severe narrowing of the aorta (A), between the pulmonary artery (P) and the superior vena cava (C). (c) In this case, the coarctation was severe: the extreme discrepancy between the size of the ductal arch (DA) and that of the hypoplastic transverse part of the arch (AO) is evident. (d) In this case of critical coarctation (tubular hypoplasia), on color Doppler, a complete inversion of the flow in the distal part of a hypoplastic aortic arch (AOA) can be seen. AD: ductal arch.

Counseling:

The overall prognosis depends on the severity of the lesion, on the presence of associated cardiac and extracardiac lesions that can significantly influence operative mortality and the life expectancy, and on correct perinatal management. The overall mortality rate is less than 5% for isolated coarctation. In symptomatic neonatal cases with associated cardiac lesions, the mortality rate is about 20%, ranging from 2% in cases associated with VSD to 40% in cases associated with complex cardiac anomalies or when the preoperative clinical condition is poor.

Ebstein's Anomaly:

Ebstein anomaly is characterized by dis-placement and attachment of one or moretricuspid leaflets (usually septal or posteriorleaflets) toward the apex of the RV. The RVis divided into an "atrialized" portion abovethe leaflets and a muscular portion below theleaflets. It accounts for less than 1% of con-genitalheartdefects,occursatarateof7%in the fetal population, and occurs in 1 per20,000 live births [1]. It is associated withmaternal lithium use, chromosomal abnormalities,ASD,patentforamenovale,andpulmonarystenosisoratresia.Ultrasoundshows apical displacement of the tricuspidvalveintotheRV,tetheredleaflets,reductionin the size of the functional RV (Fig.), in-crease in the size of the RV (including theatrialized portion), and tricuspid regurgitation.Cardiomegaly,hydrops,andtachyarrhythmias may be seen. Intrauterine mortality is as high as 85%. Differential diagnosisincludes Uhl anomaly, tricuspid valve dysplasia, and idiopathic RA enlargement [1]



Ebstein's anomaly (35 weeks of gestation). (a) Image showing the significant displacement of the tricuspid annulus within the right ventricle, with the dysplastic posterior and septal leaflets adhering to the interventricular septum (arrows). The consequent severe insufficiency is responsible for the huge cardiomegaly. (b) Color Doppler demonstrating the severe tricuspid insufficiency, with aliasing and turbulence at the level of the downwardly displaced valve (orange area).

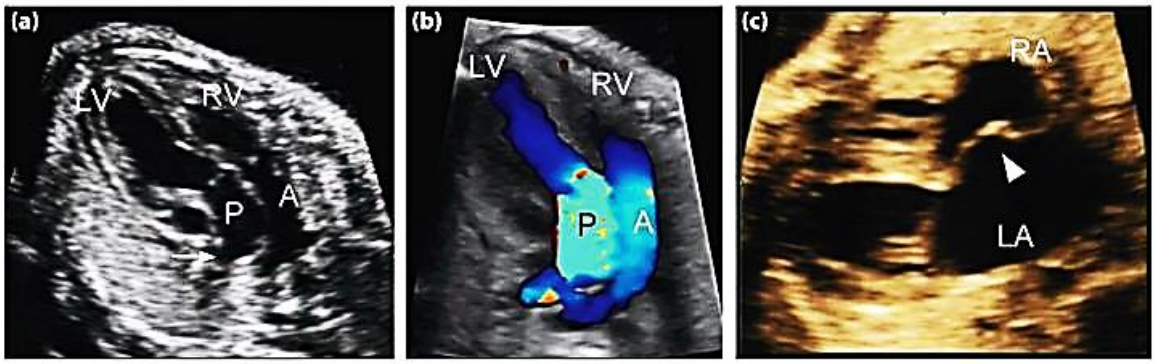
Counseling:

Defining the outcome of Ebstein's anomaly is difficult bearing in mind that there are cases who die in utero and cases who live an almost normal life. As mentioned in this section, most of the cases detected in utero are at the worse end of the spectrum, often showing severe cardiomegaly and heart

failure. This is due to the fact that only those cases in which the four-chamber view is severely abnormal are recognized in utero. In fetal series, the overall intrauterine mortality rate is as high as 40%; 20%-30% die in the neonatal period, and the remaining 30% survive more than one month. Neonates with Ebstein's anomaly requiring intensive care have a 50% mortality rate, but if they survive more than one month, then the life expectancy increases.

Transposition of Great Vessels:

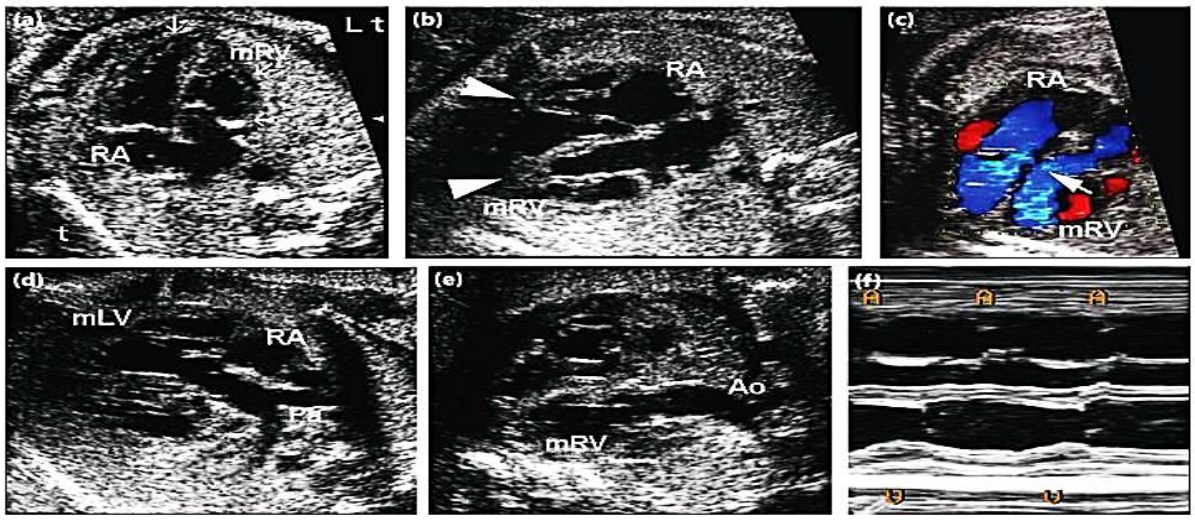
Transpositionofgreatarteriesischaracterized by the abnormal origin of the great arteries from the ventricles because of abnormal spiraling of the conotruncal septum. It isbroadly divided into D and L types. D-trans-position accounts for 80% of transpositionsand is characterized by the aorta originatingfrom the morphologic RV and the PA originatingfromthemorphologicLV.Thepulmonaryandsystemiccirculationsoperateinparallel,ratherthanserial,circuits.Oxygenationof systemic blood requires mixing via ASD,VSD, or patent ductus arteriosus. On ultra-sound, the morphologic RV is located on theright side of the morphologic LV. The arteryoriginating from the morphologic RV (i.e.,aorta)givesoffbranchestotheheadandneck,whereas the artery originating from the morphologicLV(i.e.,PA)bifurcatesandthereisasharpangleoftheleftPAwiththeductus,givingtheclassic“babybirdbeak”sign.TheaortaandPAdonotcrossbutareparalleltoeachother (Fig.), with the aorta anterior and totherightofthePA[5].Incongenitallycorrectedtransposition(L-transposition), in addition to the ventriculoarterialconcordance,thereisalsoAVdiscordance with the morphologic LA connected to the morphologic RV and the morphologic RA connected to the morphologicLV. L-transposition accounts for 1% of congenital heart defects and may be associatedwithVSDandpulmonicstenosis.Ultrasoundshows parallel aorta and PA with the aortaanterior and to the left of the PA. The tricuspid valve may be deformed and inferiorlydisplaced. Differentiating this from D-trans-position of great arteries requires identificationofthemorphologicRVandLV.



Transposition of the great arteries (TGA), 27 weeks of gestation. (a) On the left outflow tract view, the ventriculoarterial discordance and the absence of crossover (two parallel vessels) are demonstrated, with the pulmonary artery arising from the left ventricle and the anterior aorta arising from the right ventricle. (b) Color Doppler helps in defining the ventriculoarterial connection and the parallel course of the vessels. (c) In the case of TGA with an intact ventricular septum, a restrictive foramen ovale (arrowhead) indicates the need for immediate postpartum Rashkind atrioseptostomy (see this chapter's text).

Counseling:

TGA rep resents a keystone in the validation of prenatal screening of CHD, as it represents the ideal model of CHD to be screened: it has a high early neonatal mortality risk, which disappears if early neonatal management is properly planned; and, once corrected, this CHD has a greater than 90% long-term survival rate in good functional conditions. Once corrected, patients with TGA experience a 15-year survival rate of 86%.



Corrected transposition of the great arteries (cTGA), 31 weeks of gestation. (a) On the four-chamber view, the morphologic right ventricle (mRV) is connected with the left atrium, and, vice versa, the morphologic left ventricle is connected with the right atrium. Note the moderator band in the mRV and the Ebstein-like appearance of the left-sided tricuspid valve. (b) On the transverse four-chamber view, the differential insertion of the papillary muscles is evident: the tricuspid ones attach to the apex of the ventricle, whereas the mitral one attaches to the lateral free ventricular wall. (c) Color Doppler is used to demonstrate a small VSD not visible on grayscale ultrasound (arrow). (d) The right outflow tract view demonstrates the pulmonary artery (with the bifurcation) arising from the right-sided left ventricle. (e) The left outflow tract view demonstrates the aorta arising from the left-sided mRV. (f) A complete heart block can sometimes appear in the third trimester. Note the dissociation between the atrial (A) and ventricular (V) frequencies.

Counseling:

The overall prognosis of cTGA is one of the most controversial and debated issues in pediatric cardiology. The frequency and severity of the associated cardiac lesions represent the most important determinants of

survival and mortality. In particular, severe insufficiency of the left-sided tricuspid valve and impaired systolic function of the right (systemic) ventricle are the main indicators of a poor prognosis.

MRI:

The role of fetal MRI as a complementary tool to ultrasound in fetal imaging hasgrownexponentiallysincethefirstreportsin 1985 [23, 24]. Unlike ultrasound imaging,this modality is not affected by maternal andfetal conditions such as obesity and oligohydramnios [25], which particularly impairsonographic visualization of the fetal heart;maternal obesity increases the rate of sub-optimal ultrasound visualization of the fetalcardiac structures by 49.8% [26], despite advanced ultrasoundequipment.Recent studies have described the potential role of fetal MRI in the evaluation of theanatomy and pathology of the cardiovascular system [27, 28]; however, most studies asof yet had only healthy fetuses in their studypopulation. The most efficient technique tocharacterize the fetal cardiac anatomy, ac-cording to the literature, appears to be T2-weighted true fast imaging with steady-stateprecession sequence, whereas the most suit-able technique to evaluate the fetal cardiacfunctioniswithreal-timecine-MRI.

Conclusion

Routinefetalcardiacultrasoundusingfour-chamber and outflow-tract views enables thedetection and characterization of most of thecardiac anomalies. A further comprehensiveevaluation can be performed with fetal echocardiography, particularly in high-risk pregnancies and extracardiac anomalies. Dopplerimaging is used in the evaluation of vascular and valvular lesions. Three-dimensionalimaging enables reconstruction of multiplecomplex planes from a single transverse acquisition. Four-dimensional imaging enablescineloopingofimagesinmultipleplanes,enablingestimationofcardiacmotionandfunction. MRI is a complementary tool, especiallywhenfetalcardiacstructuresarevisualized sub optimallywithultrasound.

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