SOCIETY OF FETAL MEDICINE

FETAL CARDIAC ANOMALIES

A SHORT GUIDE

Fetal Cardiology Conference

SFM KARNATAKA CHAPTER

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FETAL CARDIAC ANOMALIES

A Short Guide

MESSAGE FROM MENTOR EMERITUS

The fetal heart today has shot into prominence because congenital heart disease can be diagnosed prenatally in a large number of fetuses & because these fetuses will benefit from a transfer for delivery in a tertiary care center. Additionally, using a protocol enhances the detection rate in high risk and in low risk groups. Imaging has also made great strides and pattern recognition by non-experts is becoming increasingly routine.

It is, therefore, undoubtedly with great pleasure that I write these notes on this e-monograph which will be released on the occasion of the Society of Fetal Medicine Karnataka Chapter Fetal Cardiology Conference 2023.

This document fulfills a long felt need of all stakeholders in the field of Fetal Medicine and I congratulate the entire team for this. It is a job truly well done and a very handy guide for a daily practice of fetal imaging and phenotyping.

Ashok Khurana
Mentor Emeritus
Society of Fetal Medicine

MESSAGE FROM THE PRESIDENT

Dear friends,

I am pleased to introduce a much-needed resource for fetal ultrasound examinations—**Fetal cardiac anomalies: a short guide.** For years, practitioners have desired a ready-reckoner that provides an overview of various fetal cardiac anomalies, their imaging features, and differential diagnoses. I extend my heartfelt congratulations to the Karnataka state chapter of the Society of Fetal Medicine for undertaking this challenge and creating such an invaluable academic resource.

Led by Dr. Adi Narayan Makam, the editorial team has meticulously compiled information from reputed academic sources. In this volume, they have provided a succinct summary of the common major cardiac defects, presenting the content in a clear and accessible format. It is particularly heartening to acknowledge that all the cases in this booklet have been contributed by our esteemed members from across the country, a testament to the outstanding work being conducted within our community. This successful collaboration bodes well for the academic ethos of the Society of Fetal Medicine.

I am confident that this booklet will serve as a solid foundation for practitioners venturing into the intricate world of fetal heart evaluation. Furthermore, I hope to witness the continued spirit of collaboration among our members, transforming this journal from a handy reference into a comprehensive textbook.

Warm regards,

Dr Bimal Sahani
National President
Society of Fetal Medicine

MESSAGE FROM THE PRESIDENT (KARNATAKA CHAPTER)

Dear friends and colleagues,

The heart is a remarkable organ, characterized by its proportions and the harmonious flow of blood from one chamber to another, orchestrating the symphony of life. However, this delicate balance can sometimes be disrupted during fetal development. As congenital heart defects are the most common fetal anomalies, it is imperative to include fetal cardiac examination as part of a comprehensive fetal evaluation. Mastering fetal heart evaluation requires adherence to three essential principles: Practice, Practice, and Practice.

Those performing fetal scans must familiarize themselves with the basic and extended cardiac views. Once proficiency is achieved in obtaining these views, the ability to discern the proportions of different cardiac chambers and vessels becomes second nature. An astute observer can easily identify any discrepancies, prompting a more detailed and focused evaluation.

Equally important is the skill of pattern recognition for common fetal cardiac anomalies, as it enables early detection of various cardiac defects. **"Fetal Cardiac Anomalies: A Short Guide"** serves as an atlas for these common cardiac anomalies, as well as some rare ones. The journal outlines ultrasound features of these defects, provides close differential diagnoses, and highlights additional considerations when such anomalies are encountered. I extend my sincere appreciation to the editorial team for their diligent efforts in collecting and organizing this valuable material into easily digestible nuggets of information. It is my hope that this journal will prove beneficial not only to beginners but also to seasoned practitioners as a convenient reference.

I would like to express my gratitude to all the members of the Karnataka chapter of SFM. Your whole-hearted participation in the chapter's activities inspires us to envision new academic programs. My heartfelt thanks go to the executive team of the Karnataka state chapter. Each of you is an invaluable asset, and I am grateful for your unwavering support. I also extend my appreciation to the national team of the Society of Fetal Medicine, led by our esteemed national president, **Dr. Bimal Sahani**. Their unwavering support for the academic endeavors of the Karnataka state chapter is deeply appreciated. Last but not least, I must express my profound gratitude to our mentor emeritus, **Dr. Ashok Khurana**. Without his vision, grace, and unwavering dedication, our organization would not have attained its current stature.

Warm regards,

Dr Adinarayan Makam
President
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Karnataka state chapter

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FOREWORD

Fetal cardiac abnormalities present a significant challenge for those conducting prenatal ultrasounds, given the broad range of anomalies encountered. For beginners, the task of fetal cardiac imaging can be intimidating. In response to this, we have developed a concise guide to help familiarize fetal imaging specialists with various cardiac anomalies, their common ultrasound features, and their associations. It is important to note that this guide is not intended to replace textbooks or comprehensive journal articles, but rather to serve as a quick reference and aid in becoming acquainted with different cardiac conditions.

We have made a conscious effort to include only the most important and relevant points in this short guide, ensuring its accessibility and usefulness. Our aim is to provide readers with a handy handbook that can be consulted for quick reference when needed.

It is also essential to acknowledge the collaborative nature of this journal, as it showcases the combined efforts of the members of the Society of Fetal Medicine. Their contributions, including images and cases, have been instrumental in providing a comprehensive depiction of the wide spectrum of cardiac anomalies. However, we consider this journal to be just the beginning. Our vision is to expand its content to include more cardiac anomalies and findings, and for that, we rely on the active participation and contributions of our members in future endeavors.

We invite and encourage you to engage with this journal by sharing your comments, questions, suggestions, and constructive criticism. Your input will help us refine and enhance future editions, ensuring that this resource remains valuable and relevant to the needs of our readership.

Sincerely,

The Editorial team

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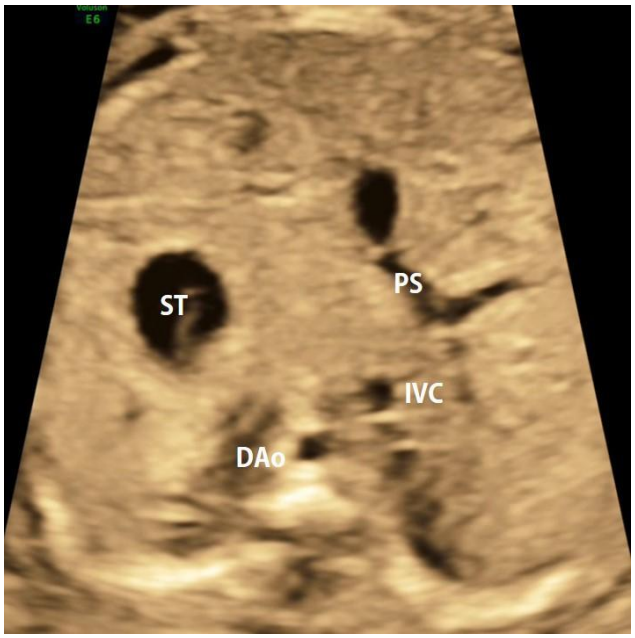
VIEWS IN
FETAL ECHOCARDIOGRAPHY

VIEWS IN FETAL ECHOCARDIOGRAPHY

Congenital heart diseases (CHD) are the leading cause of infant morbidity and mortality. A precise diagnosis of CHD in the antenatal period has proven to offer better outcome than for babies in whom the diagnosis is established in the postnatal period. Fetal echocardiography is an extension of the basic cardiac screening which is performed during routine anomaly scan. This article is a brief review of important views in the fetal cardiac exam.

UPPER ABDOMEN

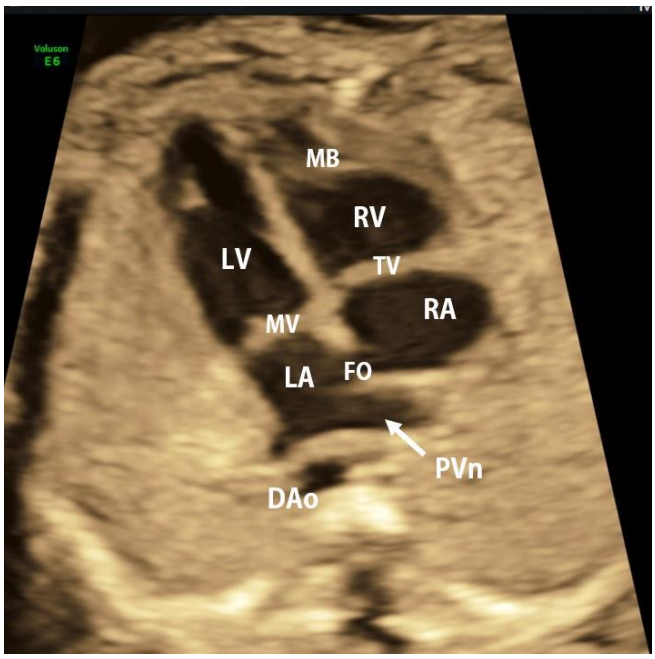


ST-Stomach
DAo- Descending aorta
IVC- Inferior Venacava
TR- Trachea
PS- Portal sinus

What to look for

- Stomach on the left side
- Descending aorta slightly towards the left of the spine
- IVC anterior and to the right of aorta
- Portal sinus turning toward the right side

FOUR CHAMBER VIEW



DAo- Descending aorta
LA- Left atrium
MV- Mitral valve
LV- Left Ventricle
MB- Moderator band
RV- Right ventricle
TV- Tricuspid Valve
RA- Right atrium
FO- Foramen Ovale
PVn- Pulmonary vein.

What to look for

Situs and general appearance

- The fetal heart occupies one-third of the thorax
- Majority of the heart in the left chest
- Cardiac axis 45 +/-20 degrees
- Cardiac apex towards the left side, formed by the left ventricle
- Four chambers are present
- No pericardial effusion
- Regular cardiac rhythm

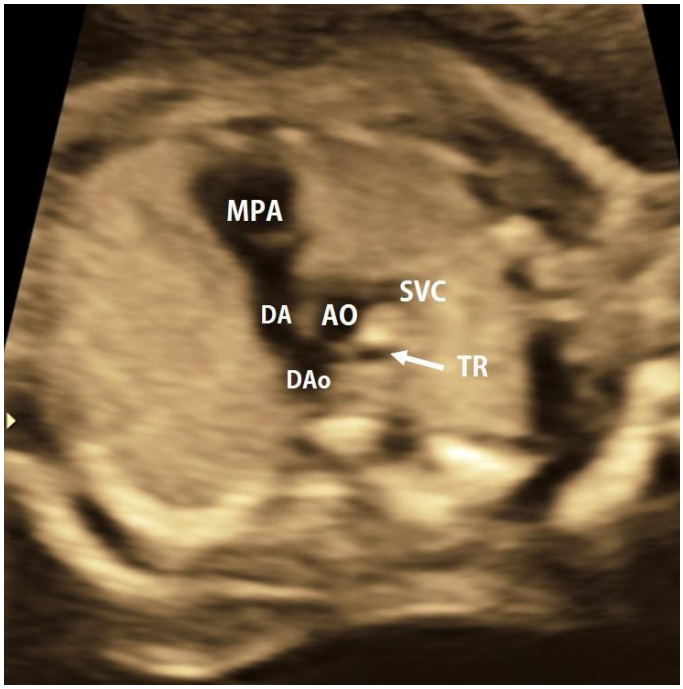
Ventricles and Interventricular septum (IVS)

- Long smooth walled left ventricle on the left
- Slightly short trabeculated right ventricle with moderator band on the right
- The tricuspid valve attached more apically than the mitral valve, forming the crux with the inlet portion of the IVS and the septum primum.
- Intact Interventricular septum.

Atria

- Two symmetrical atria
- Foramen ovale flap opening in the left atrium (LA)
- Atrial septum primum forming the crux
- Slightly rounded contour of the posterior wall of the right atrium
- Flattened posterior wall of the LA due to the entry of pulmonary veins

THREE-VESSEL TRACHEA VIEW

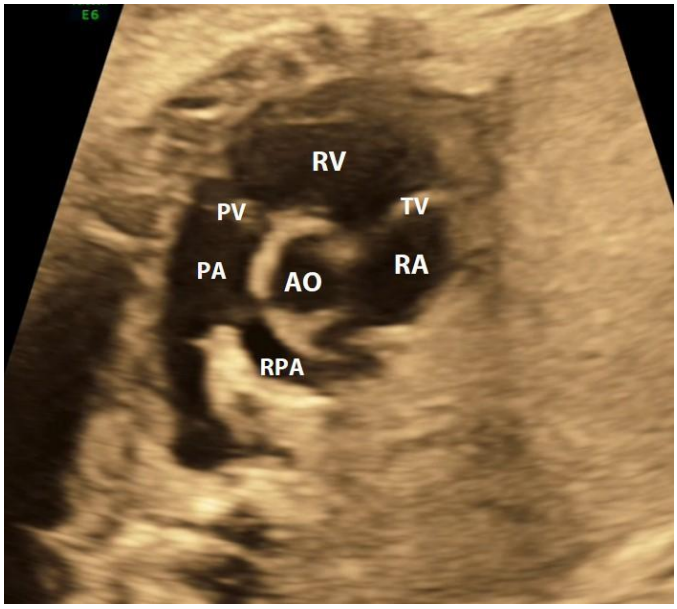


MPA- Main pulmonary artery
DA-Ductus arteriosus
DAo- Descending aorta
AO- Aorta
SVC- Superior Venacava

What to look for

- This view evaluates the pulmonary artery (PA), the aorta (AO), and the superior vena cava (SVC), arranged from left to right, anterior to posterior in orientation, and in descending order of their size
- The transverse aortic arch lies in the middle, right of the PA
- Trachea is seen as a small cystic structure with hyperechoic margins, lateral to transverse aortic arch
- The ductus arteriosus (DA) and the aortic arch form a “V” configuration to the left of the trachea
- The thymus is seen anterior to the great vessels in this view

RVOT (SHORT AXIS RIGHT VENTRICULAR OUTFLOW TRACT)



RA- Right atrium

TV- Tricuspid valve

RV- Right ventricle

PV- Pulmonary valve

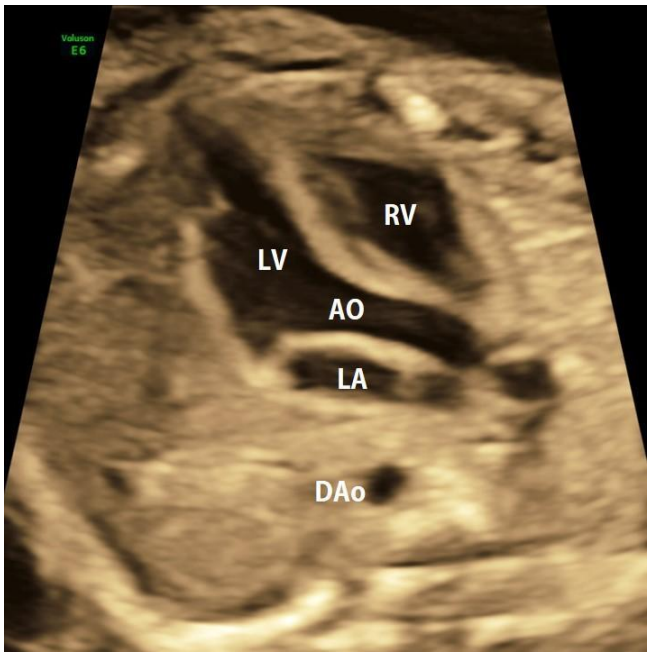
PA- Pulmonary artery

RPA- Rt pulmonary artery

What to look for

- This view depicts the entire right ventricular outflow tract wrapping around the aorta.
- The PA is seen in long axis and the aorta in cross section. This implies that the vessels are perpendicular to each other.
- PA originates from the right ventricle and branches. The right pulmonary artery (RPA) courses behind the aorta.
- This view is useful in identifying the transposition-malposition spectrum and certain ventricular septal defects (VSDs), especially the outlet type.
- The pulmonary valve is not thickened and shows normal valve excursion.

LVOT (LEFT VENTRICULAR OUTFLOW TRACT)



RV- Right ventricle

LV- Left ventricle

Ao- Aorta

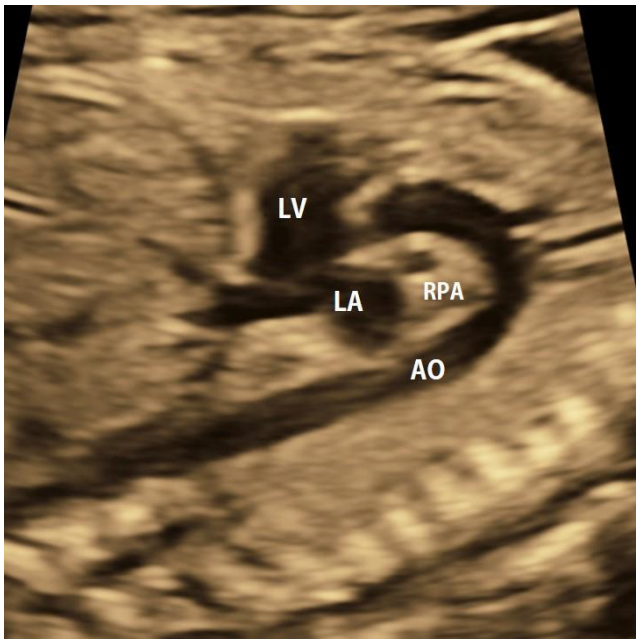
Dao- Descending aorta

LA- Left atrium

What to look for

- This view depicts the non-branching great vessel (aorta) originating from the smooth-walled left ventricle
- The IVS and the aorta are continuous, forming an angle (septo-aortic angle)
- The aorta on the other side is continuous with the mitral valve (aorto-mitral continuity).
- The aortic valve is not thickened and shows normal valve excursion.

AORTIC ARCH VIEW



LV- Left ventricle

LA- Left atrium

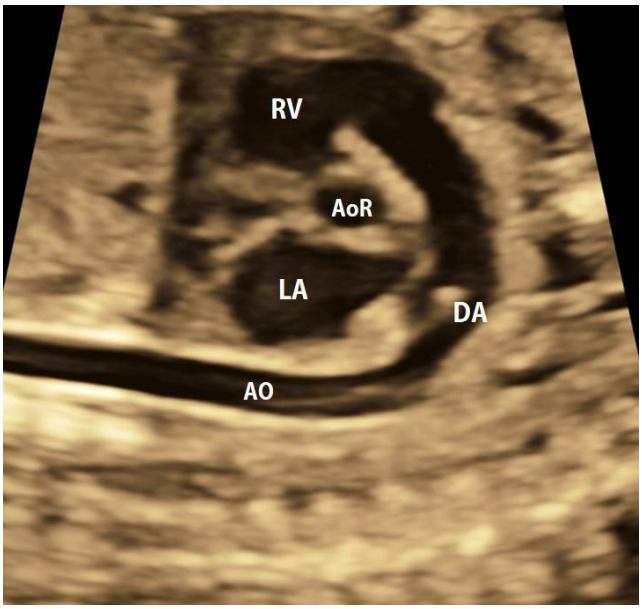
Ao- Aorta

RPA—Right pulmonary artery

What to look for

- This view depicts the aortic arch originating from the LV, from the centre of the heart.
- It has an acute curve, giving it a “walking stick” or a “candy cane” appearance.
- Three branches of the arch can be visualized in this view.

DUCTAL ARCH VIEW



RV- Right ventricle

LA- Left atrium

DA-Ductus arteriosus

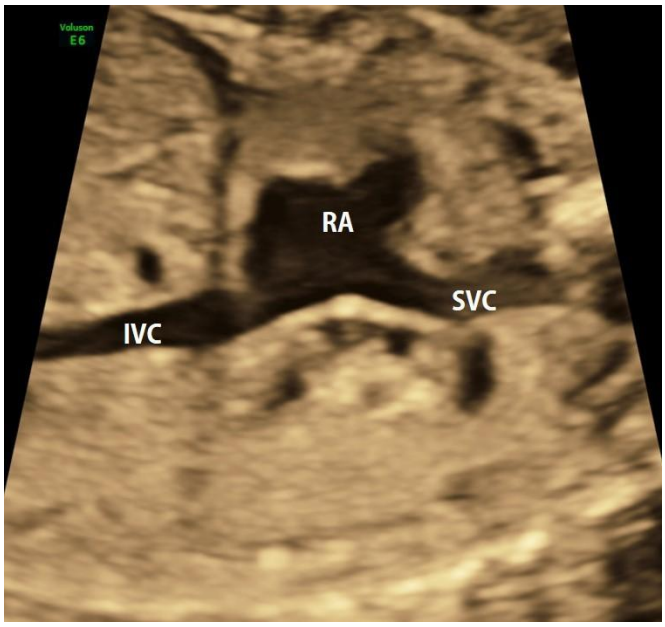
AO- Aorta

AoR- Aortic root

What to look for

- This view depicts the ductal arch originating from the RV, the anterior-most ventricle
- It has a wide curve, giving it a “hockey stick” appearance
- No branches arise from this arch

BICAVAL VIEW



SVC- Superior venacava

RA- Right atrium

IVC- Inferior venaceva

What to look for

- This view depicts the two veins, superior vena cava (SVC) and inferior vena cava (IVC), entering the right atrium
- SVC entry into the right atrium shows continuity between the posterior wall of the SVC and the interatrial septum
- The IVC at the junction with the right atrium appears wide due to the confluence of the ductus venosus and the hepatic veins

FIRST TRIMESTER

Improved ultrasound technology and the advent of new-generation transducers have enabled the visualization of the heart in early gestation. It is better to evaluate the heart after 12 weeks (3 days) for a better yield of the exam.

Situs

- The position of the heart and the stomach is on the same side of the fetus

Four Chamber

- Assessed with grayscale and color mode. The use of dual mode is better if that option is available
- The cardiac axis is to be assessed in this view
- Look for symmetrical ventricular inflow

Three-vessel-trachea view (3VT)

- Color Doppler is applied. Look for forward flow in both the great vessels
- Formation of a "V" to the left of the trachea

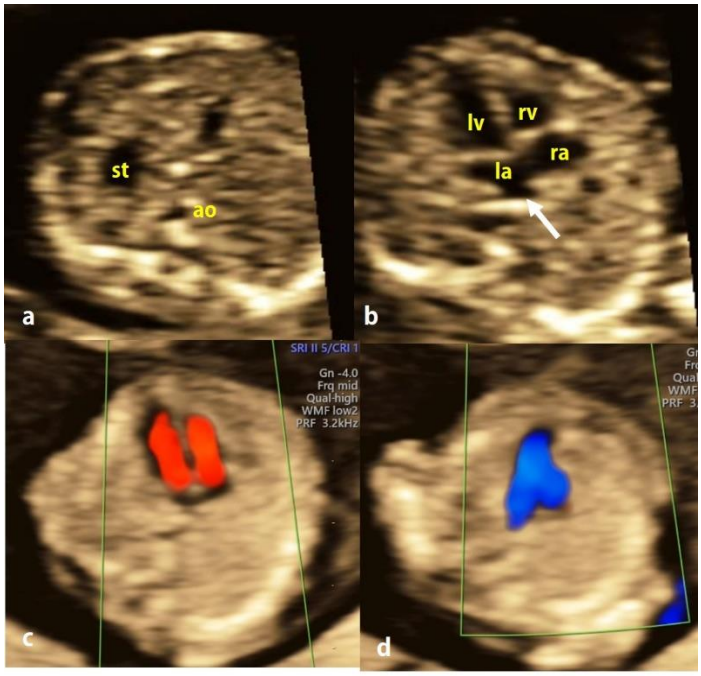


Fig.1: (a) Stomach and aorta on the left side(b) Four-chamber view shows normal cardiac axis, four distinct chambers, AV valves, and pulmonary veins (arrow) (c) colour Doppler shows symmetrical ventricular inflow (d). Three-vessel-trachea view shows forward flow in both outflows.

Optional views

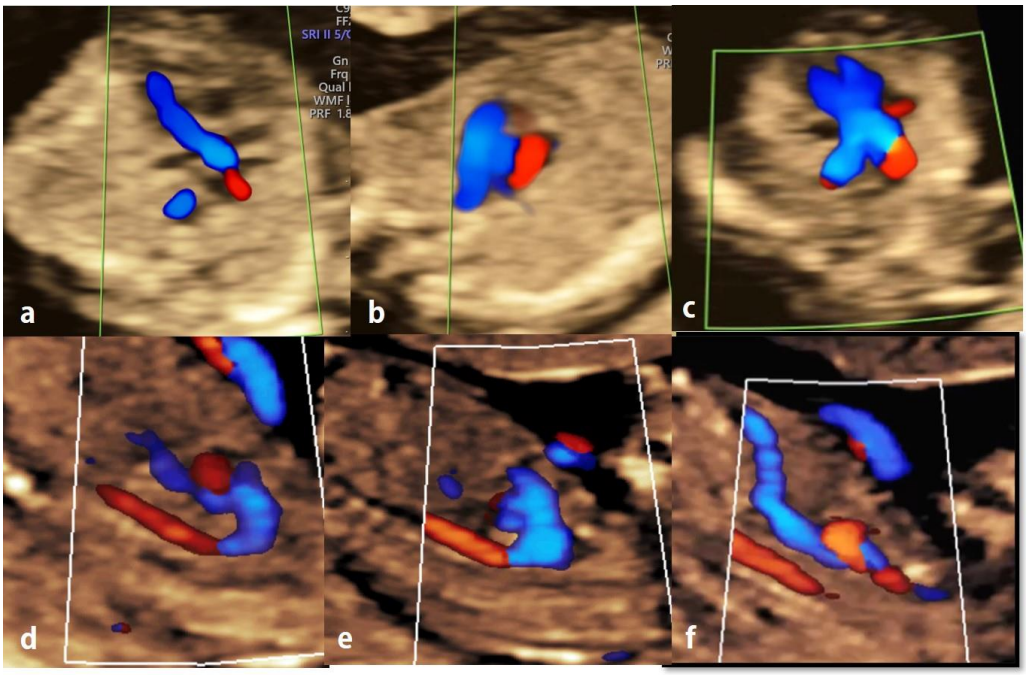


Fig.2: (a) LVOT (b). RVOT (c).Crossing of vessels-"X" sign. (d) Aortic arch (e) Ductal arch (f) Bicaval view

- Outflow tract and arch views are challenging. These can be tried if there is a suspicion of CHD in the primary views. In such cases, the patient should be called back for an early fetal echo

Suggested reading

- ISUOG Practice Guidelines (updated): Fetal cardiac screening: Ultrasound Obstet Gynecol 2023; 61: 788–803.
- AIUM Practice Parameter for the Performance of Fetal Echocardiography, J Ultrasound Med 2020; 39:E5–E16.
- A Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts. 4th edition. A. Abuhamad.,R. Chaoui.

SEPTAL DEFECT

VENTRICULAR SEPTAL DEFECT

The most frequent congenital heart anomalies are ventricular septal defects (VSD), which result in hemodynamic connections between the left and right ventricles. These can occur either in isolation or in conjunction with other cardiac abnormalities. In some cardiac abnormalities, VSD is necessary (i.e., obligatory). There are four types of VSDs: inlet, outlet, muscular, and perimembranous.

ULTRASOUND FEATURES

- General features of VSDs include focal interruption in the interventricular septum (IVS), buckling of IVS and bright echogenic margins of the defect.
- **Muscular VSD -** A defect in the muscular portion of the septum (Fig. 1), which extends from the attachment of the tricuspid valve to the apex of the heart. Best seen in an apical or lateral four-chamber view. Small defects may be detected only with color Doppler. Bidirectional flow is commonly noted. If unidirectional flow is observed, aortic or pulmonary valve stenosis should be ruled out.
- **Inlet VSD -** A defect in the part of the IVS septum that lies between the tricuspid and the mitral valves (Fig.2). An inlet VSD can be a part of an atrioventricular septal defect (AVSD). Best seen in the four-chamber view in the plane of the atrioventricular valves.
- **Perimembranous VSD -** A defect in the small, thin membranous region of the left outflow tract, just beneath the aortic valve and crista supraventricularis (Fig.3). Best seen in the five-chamber view or LVOT view as an interruption in the septo-aortic continuity.
- **Outlet VSD –** A defect in the septum involving the conal and infundibular regions, below the arterial valves, and above the crista supraventricularis. These are rare and difficult to detect prenatally. Best seen in a plane slightly cranial to the five-chamber view and in short axis views at the level of the outflow tracts.

DIFFERENTIAL DIAGNOSIS

- Dropout artifact, mainly in the perimembranous region – No echogenic margins, septal continuity is present in lateral four-chamber and LVOT views.

ADDITIONAL POINTS

- Obligatory association with Atrioventricular Septal Defect (AVSD), Tricuspid Atresia with VSD, Mitral Atresia with VSD, Double Inlet Mono ventricle, Double Inlet Left Ventricle (DILV), Pulmonary Atresia with VSD, Absent Pulmonary Valve Syndrome (APVS), Common Arterial Trunk (CAT), Double Outlet Right Ventricle (DORV) and type 2 Interrupted Aortic Arch.
- Occasional association with D-transposition of Great Arteries (d-TGA), Corrected Transposition of Great Arteries (ccTGA) and Aortic Coarctation.
- VSDs are never associated with hypoplastic left heart syndrome (HLHS). If there is a VSD, HLHS should not be the diagnosis.

- The prognosis of a VSD is dependent on the size and location of the VSD and associated cardiac and extracardiac malformations.
- Small (less than 3 mm) muscular and small perimembranous (less than 50% of the size of the aortic valve diameter) have an excellent outcome; up to 80% close spontaneously by birth or in the first 2 years of life.
- Moderate-to-large-sized VSDs may necessitate surgical closure in order to reduce long term morbidities.
- When a perimembranous VSD is detected, a detailed assessment of the great arteries is critical, given the strong association with conotruncal abnormalities.
- Chromosomal abnormalities have been reported in more than 20% of fetuses with VSD, commonly in association with other cardiac malformations.
- Isolated muscular VSDs diagnosed prenatally have no increased risk of chromosomal abnormalities.
- VSDs are the most common lesion in many chromosomal abnormalities, such as trisomy 21, 18, and 13.
- VSDs, in combination with other extracardiac anomalies, have a high risk of the presence of pathogenic copy number variants (CNVs).

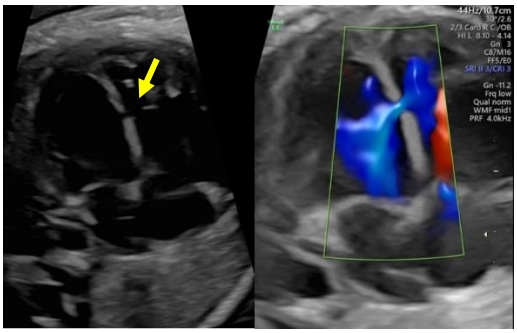


Fig. 1: Muscular VSD (arrow). A defect is seen in the muscular septum

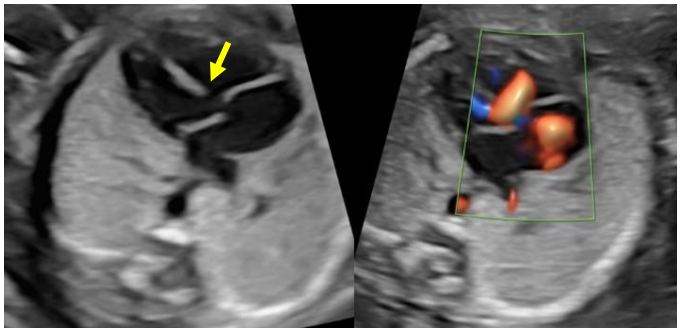


Fig. 2: Inlet VSD (arrow). A defect is seen at the level of the atrioventricular valves

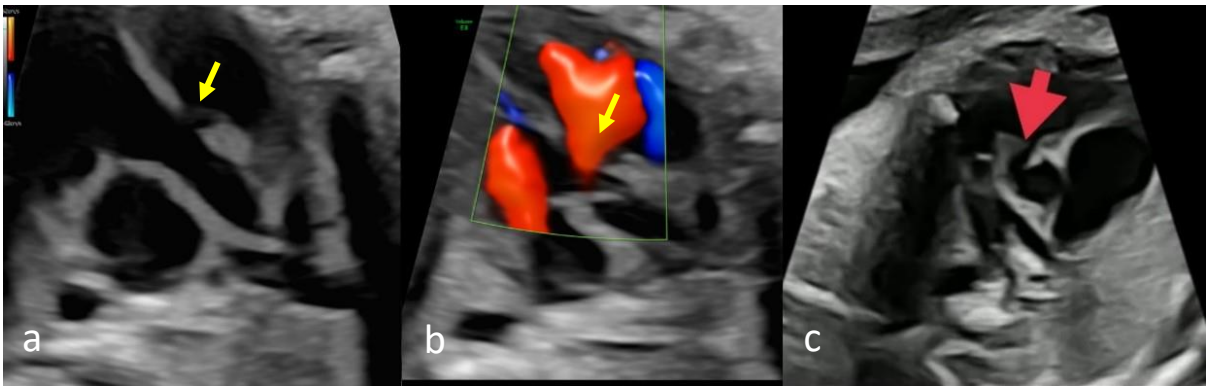


Fig. 3: Perimembranous VSD (a) A defect is seen in the LVOT view as septo-aortic discontinuity (arrow) (b) Flow across the defect (arrow) (c) Short axis view showing perimembranous VSD, seen as a defect in the aortic ring (pink arrow)

ATRIOVENTRICULAR SEPTAL DEFECT

Atrioventricular septal defect (AVSD) is a spectrum of cardiac malformations in which there is a single atrioventricular (AV) valve opening into both ventricular chambers. AVSD is also called an AV canal defect, common AV canal, or endocardial cushion defect.

AVSD is classified as complete or partial. **Complete AVSD** has defects in the atrial and ventricular septum immediately above and below the common AV valve. A **partial defect** has a defect in the atrial septum primum, accompanied by a cleft in the mitral valve.

AVSD is also classified as balanced or unbalanced. When blood drains both ventricles almost equally in diastole, this is called a **balanced AVSD.** However, when there is unequal drainage (often due to right-or-left outflow obstruction), this results in a disproportion between the two ventricles and is called an **unbalanced AVSD.**

ULTRASOUND FEATURES

- A four-chamber view shows a linear insertion of the AV valves (Figs. 1a, 2). In a normal heart, the tricuspid valve is more apically inserted than the mitral valve (AV valve offset); loss of offset indicates a common AV valve. This is best seen in an apical four-chamber view in systole.
- In **complete AVSD**, there are defects in the atrial and ventricular septum. This is seen as a defect in the center of the heart in diastole due to lack of the crux (Fig.1b).
- Color Doppler shows a single channel of blood during diastole that divides over the remnant of the ventricular septum (Fig.2a). There is often regurgitation jet of common valve.
- In **partial AVSD**, there is a defect in the septum primum of the atrium (the part of the atrial septum near the crux), but there is no large ventricular septal defect (Fig. 2b).
- In a **balanced AVSD**, both ventricles are equal in size.
- In **unbalanced AVSD**, there is a disproportion in the size of the ventricles (Fig. 2c). This is accompanied by one of the outflow tracts being of smaller caliber, indicating obstruction or stenosis.

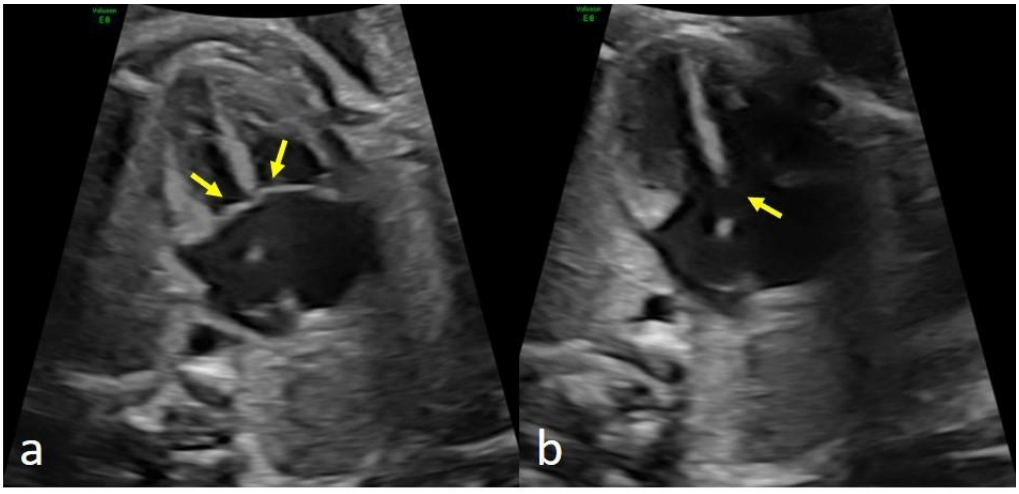


Fig. 1: Complete balanced AVSD (a) Equal sized ventricles. Linear insertion of AV valves (arrows) (b) In diastole, a hole in the center of the heart is seen (arrow) due to absent crux

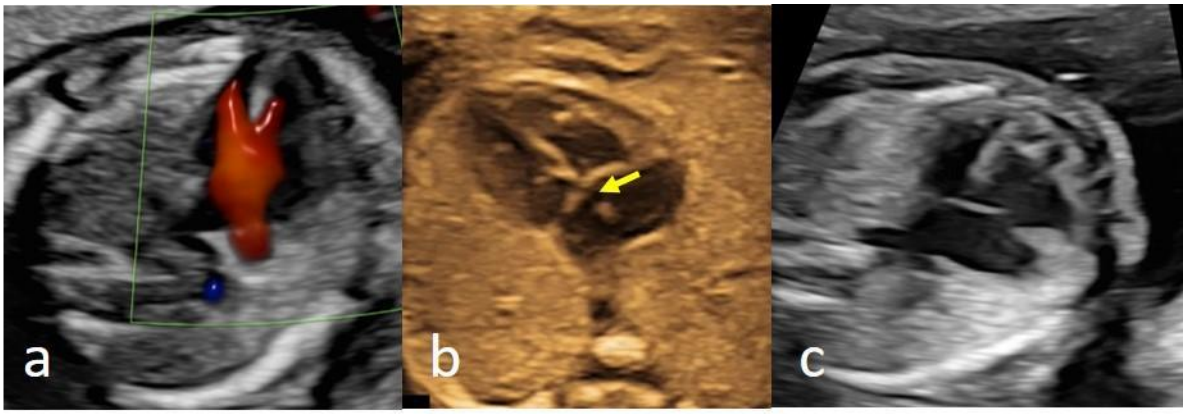


Fig. 2: (a) Color Doppler showing a single channel of blood during diastole that divides over the remnant of the ventricular septum in a case of AVSD (b) Partial AVSD; linear insertion of AV valves; a small defect in the septum primum of the atrium (arrow) (c) Unbalanced AVSD; linear insertion of the AV valves; and ventricular asymmetry

DIFFERENTIAL DIAGNOSIS

- Isolated inlet ventricular septal defect (VSD)
- Dilated coronary sinus, in cases of persistent left superior vena cava (PLSVC)

ADDITIONAL POINTS

- The atrioventricular length ratio (AVL ratio) can be used to facilitate the detection of AVSD. In normal hearts, the atria are half the length of the ventricles (AVL ratio 0.5). In fetal hearts with AVSD, the atria appear larger; hence, the ratio of the atrial length and the ventricular length is increased. A cut-off value of the AVL ratio > 0.6 detects AVSD in 83% of cases, with a false-positive rate of 5.7%.
- Situs must be carefully evaluated, as many cases of AVSD are associated with heterotaxy.
- Associated cardiac defects include tetralogy of Fallot, double outlet ventricle, right aortic arch, and other conotruncal anomalies. Pulmonary atresia and anomalies of the pulmonary and systemic veins can also be found, mainly in association with both left and right isomerism.
- Isolated AVSD is associated with trisomy 21 in 58% of cases.
- In AVSD with atrial isomerism, the fetal karyotype is usually normal.

LEFT HEART ANOMALIES

HYPOPLASTIC LEFT HEART SYNDROME

A range of cardiac abnormalities known as hypoplastic left heart syndrome (HLHS) have a significantly underdeveloped left ventricle (LV) and left ventricular outflow tract, thus obstructing the systemic cardiac output.

ULTRASOUND FEATURES

Two main forms of HLHS are observed:

- HLHS caused by severe aortic stenosis or aortic atresia: this results in a visible LV with a globular shape and poor contractility. The inner wall of the LV appears brightly echogenic due to **endocardial fibroelastosis**. The mitral valve is patent but usually severely dysplastic.
- HLHS due to atresia of both mitral and aortic valves: this results in practically no communication between the left atrium (LA) and the LV. Hence, the LV is nearly absent or severely hypoplastic (Fig.1). There is no demonstrable flow from the LA to the LV on pulsed-wave or colour Doppler.
- The cardiac apex is predominantly formed by the right ventricle (RV) (Figs.1 and 2).
- The left atrium is small. There is a reversed flow from left to right across the foramen ovale.
- In the five-chamber view, the hypoplastic aortic outflow is difficult to visualise (size <3 mm).
- Reversed flow on pulse-wave or color Doppler in the aortic arch (Fig. 2c). The ductus arteriosus retrogradely perfuses the coronary arteries and the head and neck vessels.

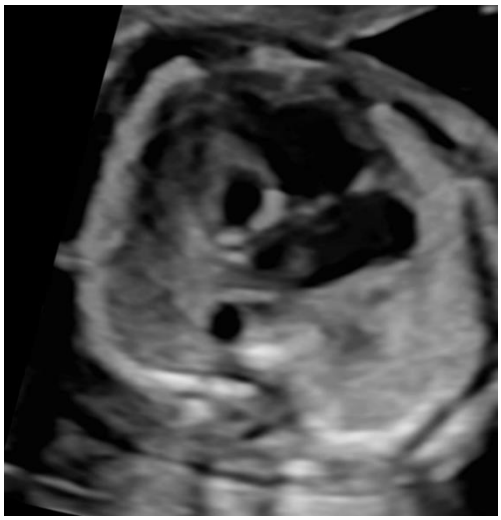


Fig.1: Small, thick-walled left ventricle in a case of HLHS. Cardiac apex is formed by the right ventricle.

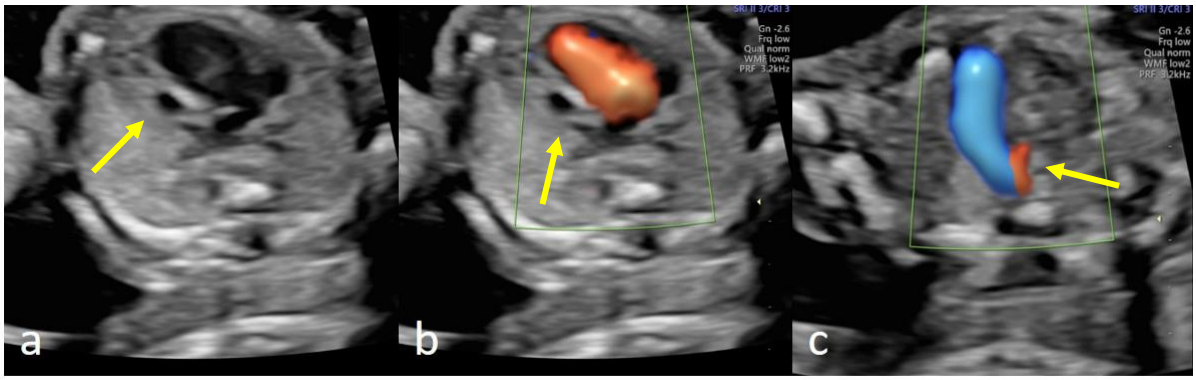


Fig.2: HLHS (a) Four chamber view: shows small sized LV with bright echogenic inner wall (arrow). Cardiac apex formed by the RV. (b) No flow across the mitral valve (arrow) (c) Three-vessel-trachea view showing retrograde flow in aortic arch (arrow).

DIFFERENTIAL DIAGNOSIS

- Coarctation of Aorta - narrow aortic arch with predominantly forward flows.
- Mitral Atresia with VSD - no flow across the mitral valve. However, the left ventricular cavity appears patent as it is perfused from the right ventricle via a VSD.
- Unbalanced atrioventricular septal defect - linear insertion of the atrioventricular valves.
- Critical Aortic Valve Stenosis - the aortic outflow is patent, shows forward flow with severely elevated flow velocities due to severe stenosis.

ADDITIONAL POINTS

- There is no ventricular septal defect in HLHS. If VSD is present, HLHS should not be the diagnosis.
- Left ventriculo-coronary arterial communications (VCACs) may sometimes be seen (Fig. 3).
- HLHS can be associated with chromosomal abnormalities in 4–5% of cases, e.g., Turner Syndrome, Trisomy 13, 18, and others.

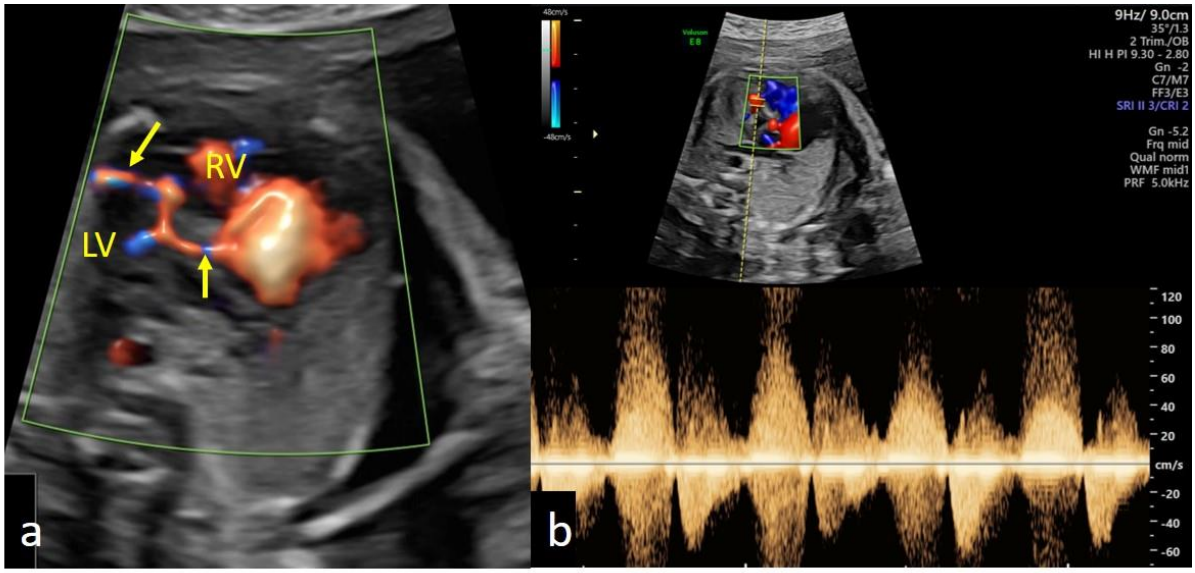


Fig.3: Ventriculo-coronary communications in HLHS (a) HD-color flow shows a dilated coronary artery and its branches (arrows) in the interventricular septum due to ventriculo-coronary arterial communications (VCAC) (b) Pulse-wave Doppler shows a typical bi-directional flow in the VCAC

AORTIC STENOSIS

Aortic stenosis (AS) is defined as obstruction of the left ventricular outflow due to narrowing at the level of the aortic valve. Valvular aortic stenosis is the most common type and can be diagnosed prenatally. Rarely, the stenosis may be subvalvular or supravalvular; accurate prenatal detection of these is difficult.

ULTRASOUND FEATURES OF MILD AORTIC STENOSIS

- Four-chamber view is often normal in mild aortic stenosis.
- Five-chamber view may show post stenotic dilation of the ascending aorta (Fig. 1 a).
- Thickened aortic valve leaflets, doming, and a lack of complete opening of the valve may be seen.
- A cross section at the level of the aortic valve may show a bicuspid aortic valve and/or thickening of the cusps.
- Most cases are detected on color Doppler, which shows turbulent, antegrade flows across the aortic valve and the aortic arch (Fig.1 b).
- On spectral Doppler, peak systolic velocities of120 to 150 cm/sec are seen, often with slow acceleration to peak velocity.

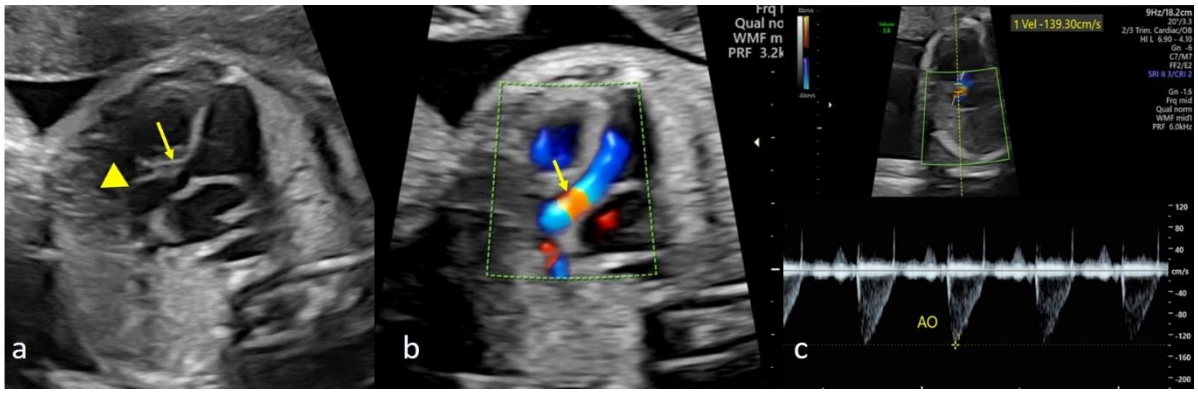


Fig.1: Mild aortic stenosis (a) Mildly narrow aortic root (arrow), thickened aortic valve, and post stenotic dilatation (arrow head) (b) Color Doppler - aliasing across the aortic valve (arrow) (c) Spectral Doppler shows elevated PSV

ULTRASOUND FEATURES OF CRITICAL AORTIC STENOSIS

- An abnormal four-chamber view with a dilated, globular left ventricle (Fig.2a), showing reduced contractility.
- Five-chamber view shows a narrow aortic root (Fig.2 b), thickened aortic valve, and decreased movements of the aortic valve leaflets.
- Color Doppler shows highly turbulent but antegrade flows across the aortic valve (Fig. 2c).
- Three-vessel-trachea view often shows reversal of flow in the aortic arch.
- Peak systolic velocity across the aortic valve is typically >200 cm/sec

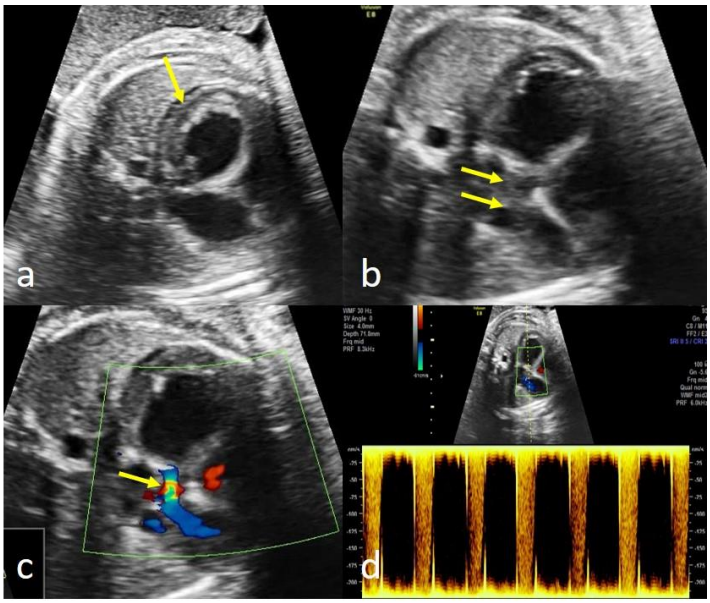


Fig.2: Critical aortic stenosis (a) Four-chamber view shows a dilated, globular left ventricle (arrow) (b) Five-chamber view shows a severely narrow aortic root (arrows) and thickened aortic valve (c) Color Doppler shows aliasing across the severely narrow aortic valve (arrow) (d) Spectral Doppler shows severely elevated PSV (>300 cm/sec)

DIFFERENTIAL DIAGNOSIS

- Hypoplastic left heart syndrome (HLHS) – The left ventricle is hypoplastic, the aortic valve is atretic, and there is no forward flow across the valve.

ADDITIONAL POINTS

- A narrow aortic root with a dilated ascending aorta should raise suspicion of aortic stenosis.
- Associated cardiac malformations include bicuspid aortic valve, ventricular septal defect (VSD) and coarctation of aorta.
- Mild aortic stenosis can rarely progress to critical aortic stenosis and HLHS.
- Aortic stenosis in association with mitral stenosis with a normally contracting left ventricle is termed the “Shone complex.”
- Aortic stenosis associated with renal anomalies, nuchal thickening, or hydrops may suggest Turner syndrome.
- Aortic stenosis is also associated with Williams–Beuren syndrome (deletion 7q11.23).

RIGHT HEART ANOMALIES

TRICUSPID ATRESIA

Tricuspid atresia is defined by a loss of the right atrioventricular connection, which prevents communication between the right atrium and the right ventricle and causes the right ventricle to shrink in size. The size of the right ventricle depends on the size of the ventricular septal defect, which is nearly always present.

ULTRASOUND FEATURES

- Four-chamber view shows a small right ventricle and a perimembranous or inlet ventricular septal defect.
- Absent right atrio-ventricular junction, seen as thickened echogenic tissue.
- No flow across the tricuspid valve.
- Interatrial septum & interventricular septum are malaligned.
- Ventriculo-arterial connections can be concordant or discordant.

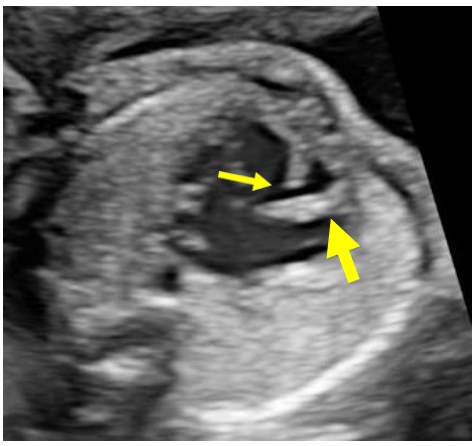


Fig.1- Four chamber view showing a small right ventricle with echogenic thickened tricuspid valve (large arrow) and an inlet VSD (small arrow) in a case of Tricuspid Atresia

DIFFERENTIAL DIAGNOSIS

- Pulmonary atresia with intact ventricular septum - thick-walled right ventricle, intact interventricular septum, concordant great vessels, with retrograde flow in the pulmonary artery.
- Double inlet left ventricle– Both atrioventricular valves are patent and drain into a common ventricle.

ADDITIONAL POINTS

- Types of TAVSD
  - Type I (70-80%) - associated with normally oriented great arteries.
  - Type II - associated with D- transposition.
  - Type III- associated with complex great vessels like Truncus arteriosus or L- transposition.
- Pulmonary or aortic stenosis is common
- Ductal arch usually shows forward flow unless associated with severe pulmonary stenosis.

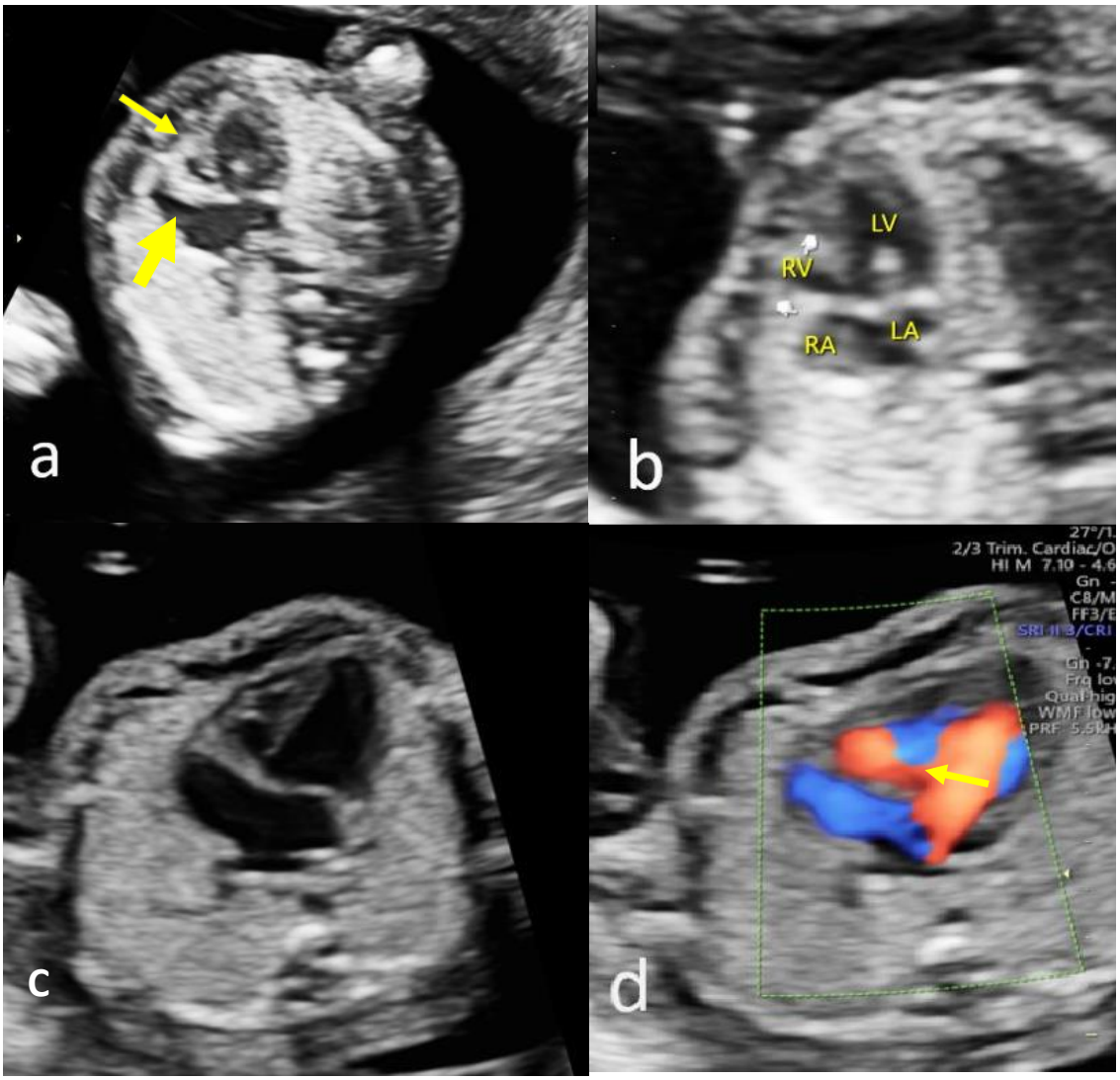


Fig 2: (a) Four chamber view at 13 weeks shows a small right ventricle (small arrow) with an echogenic tricuspid AV valve (large arrow). (b & c) Four -chamber view confirming the same findings at 16 weeks and 22 weeks (d) Color Doppler shows no flow across the tricuspid valve. The right ventricle is filling via a VSD (arrow).

EBSTEIN ANOMALY

In a normal heart, the tricuspid valve inserts slightly more apically on the interventricular septum than the mitral valve. In Ebstein anomaly (EA), the septal and posterior leaflets of the tricuspid valve are displaced inferiorly from the tricuspid valve annulus towards the apex of the heart and originate from the right ventricular myocardium. The anterior tricuspid leaflet maintains its normal attachment to the tricuspid valve annulus. The proximal portion of the right ventricle is then continuous with the true right atrium and forms an "atrialized" portion of the right ventricle (RV). EA shows a wide spectrum. Minor forms may show minimal displacement of the tricuspid valves and mild tricuspid regurgitation; severe forms show atrialization of the entire RV and severe tricuspid regurgitation.

ULTRASOUND FEATURES

- Cardiomegaly with increased cardiothoracic ratio.
- Dilated right atrium (Fig.1).
- Increase in atrioventricular valve offset - the septal leaflet of the tricuspid valve is displaced inferiorly towards the cardiac apex of the heart (Fig.1).
- Color Doppler shows tricuspid valve regurgitation during the entire systole (holosystolic) (Fig.2). The origin of the regurgitant jet is deep within the RV.
- Color Doppler of the right outflow tract shows reverse flow in the ductus arteriosus towards the pulmonary valve when associated with pulmonary atresia (Fig.2 e) or antegrade flow into the typically narrow pulmonary trunk when associated with stenosis.
- In severe EA with a large atrialized ventricle, paradoxical movements of the ventricular septum can be observed, with the apical and basal parts of the interventricular septum moving in opposite directions.

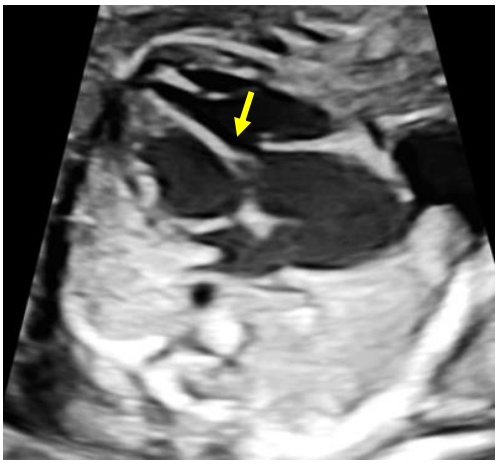


Fig.1: Ebstein anomaly. The right atrium is dilated. The septal leaflet of the tricuspid valve (arrow) is displaced apically

- Severe forms may also present with pulmonary regurgitation back into the RV, thus increasing the risk of cardiac failure, hydrops, and demise.

DIFFERENTIAL DIAGNOSIS

- Tricuspid Valve Dysplasia – The septal leaflet is inserted normally at the valve annulus. The origin of tricuspid regurgitation is at the typical anatomic valve annulus.
- Premature closure of Ductus Arteriosus - Aliasing and elevated velocities in the ductus arteriosus.

ADDITIONAL POINTS

- Associated cardiac defects include pulmonary stenosis or atresia.
- Pulmonary stenosis and atresia are often functional due to reduced flow across the pulmonary valve due to severe tricuspid regurgitation.
- Postnatally, atrial dilatation due to severe tricuspid regurgitation frequently results in a secondum-type atrial septal defect. Though it is difficult to detect prenatally, an enlarged foramen ovale can be a clue.
- In severe EA, a fetal circular shunt can sometimes form, where blood from the tricuspid regurgitant flows into the left ventricle through the foramen ovale. The blood is then ejected into the aorta, goes via the ductus arteriosus, and back into the RV through pulmonary artery regurgitation. This circular flow pattern is associated with high fetal mortality.
- Significant cardiomegaly may cause secondary lung compression and consequent hypoplasia, fetal hydrops, and death.
- Recent studies have shown an increased association between trisomy 21 and EA diagnosed in early gestation.

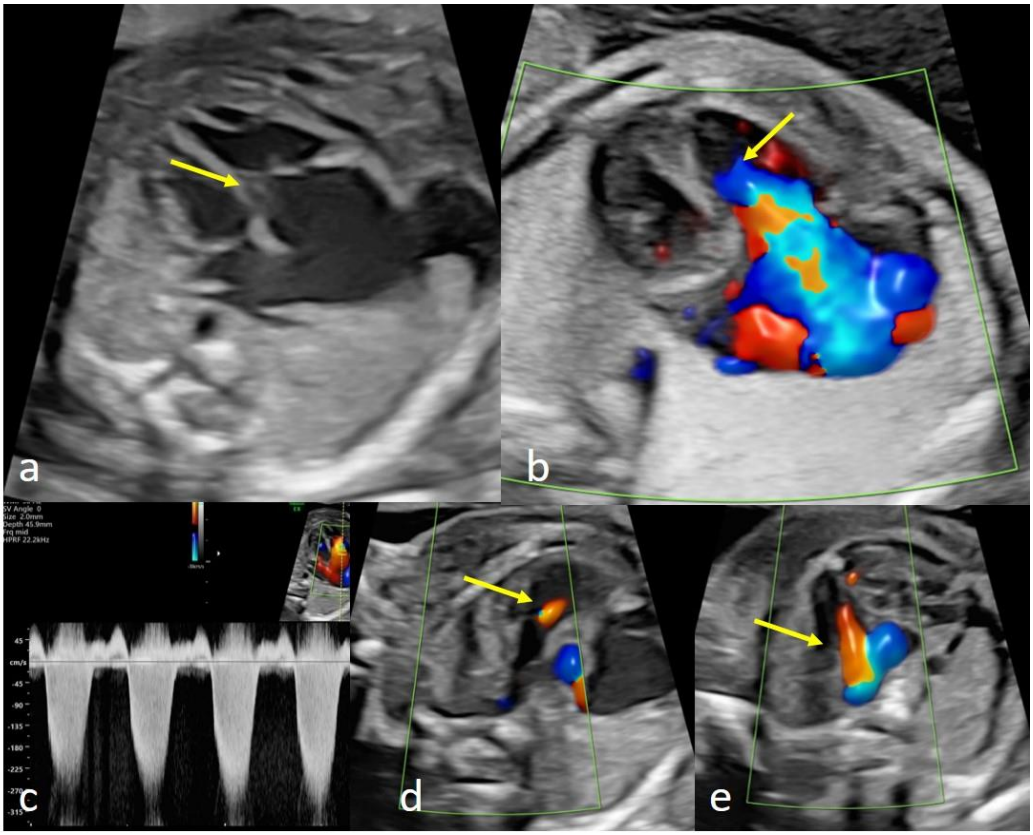


Fig.2 : Ebstein anomaly (a) Four chamber view shows cardiomegaly, enlarged atrium and more apical insertion of the septal leaflets of the tricuspid valve on the interventricular septum (arrow) (b) Color Doppler shows the origin of the severe tricuspid regurgitation jet from deep in the right ventricle (arrow) (c) Spectral Doppler shows tricuspid regurgitation velocity more than 200 cm/sec (d) Right ventricular outflow tract view shows pulmonary regurgitation (arrow) (e) Three-vessel-trachea view shows retrograde flow in the pulmonary artery (arrow)

TRICUSPID DYSPLASIA

Tricuspid dysplasia (TD) is a diverse group of anomalies that affect the tricuspid valve. The tricuspid valve shows a normal anatomic insertion of the leaflets at the tricuspid valve annulus. The spectrum ranges from minor lesions involving mild thickening of the valve leaflets to severely dysplastic valve leaflets with anomalous chordae tendineae insertion.

ULTRASOUND FEATURES

- Cardiomegaly with an enlarged right atrium (RA).
- Thickened valve leaflets that do not close properly in systole.
- Tricuspid valve regurgitation during systole. On color Doppler, the origin of the regurgitant jet is at the anatomic valve annulus.
- Color Doppler of the right outflow tract shows antegrade flow into the typically narrow and stenosed pulmonary trunk. Reverse flow in the pulmonary artery and ductus arteriosus may be seen when TD is associated with pulmonary atresia.
- Occasionally, severe forms may also present with pulmonary regurgitation back into the right ventricle, thus increasing the risk of cardiac failure, hydrops, and demise.

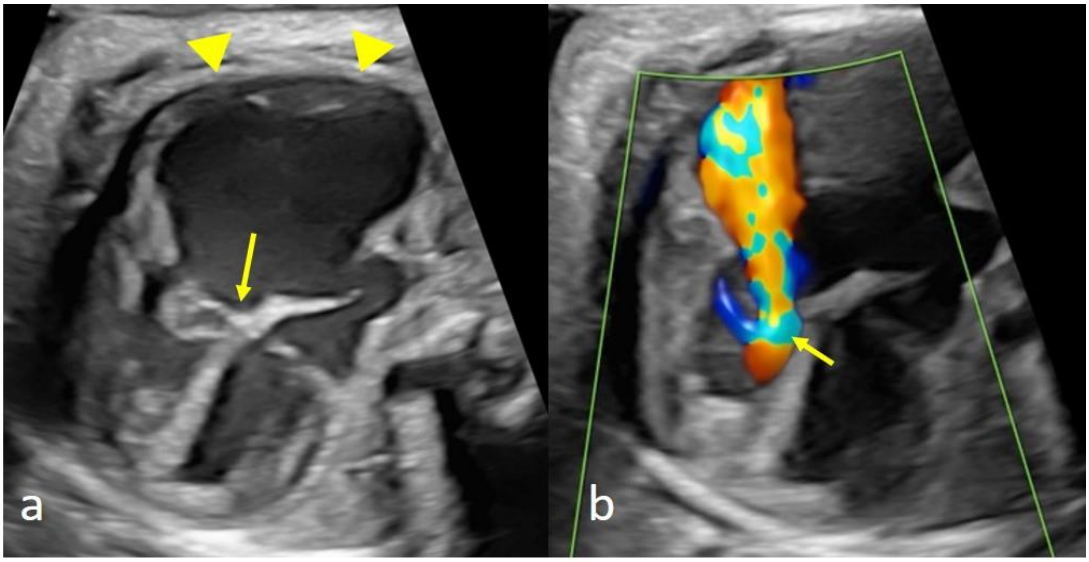


Fig.1: Tricuspid dysplasia (a) Four chamber view shows cardiomegaly, enlarged right atrium (arrowheads) and mildly thickened valve leaflets. The tricuspid valve leaflets are attached to valve annulus (arrow). (b) Color Doppler shows severe tricuspid regurgitation originating from the anatomic valve annulus (arrow)

DIFFERENTIAL DIAGNOSIS

- Ebstein Anomaly – The septal leaflet shows apical insertion towards the cardiac apex.
- Pulmonary Atresia with intact ventricular septum- Thickened right ventricular walls and no flow across the pulmonary valve
- Premature closure of Ductus Arteriosus – aliasing and high velocities in the ductal arch.

ADDITIONAL POINTS

- Associated cardiac defects include pulmonary stenosis, pulmonary atresia and atrial septal defect.
- Pulmonary stenosis and atresia are often functional due to reduced flow across the pulmonary valve due to severe tricuspid regurgitation.
- TD is commonly associated with chromosomal anomalies, mainly Trisomy 18.

PULMONARY ATRESIA WITH INTACT VENTRICULAR SEPTUM

Pulmonary atresia with intact ventricular septum (PAIVS) is a group of cardiac malformations that have absent communication between the right ventricle (RV) and the pulmonary arterial circulation, in combination with an intact interventricular septum.

ULTRASOUND FEATURES

- Hypokinetic right ventricle with thickened walls (Fig.1).
- The size of RV can vary from absent RV to normal sized ventricle, but the contractility of RV is significantly reduced.
- Tricuspid valve is patent, though often dysplastic with a narrow annulus and abnormal excursion of the leaflets.
- Progressive hypertrophy of the RV walls with advancing gestational age and hypertrophy of the ventricular septum may lead to a bulge into the left ventricle (LV).

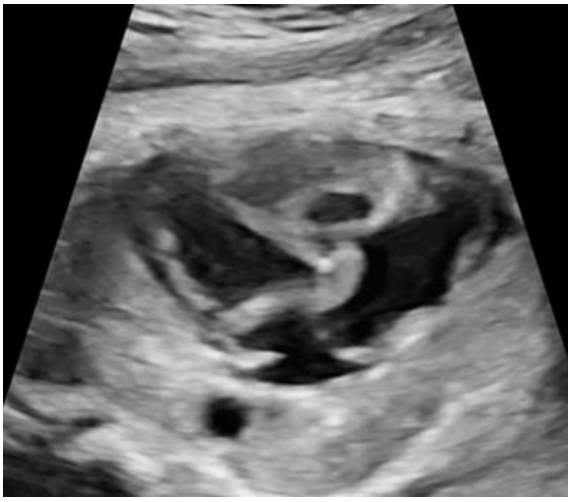


Fig.1: PAIVS. Thickened RV walls and intact ventricular septum

- The main pulmonary artery in the right outflow tract view or the three-vessel view is typically small or hypoplastic. In severe cases, the pulmonary artery is not visible on gray scale and is seen only with color Doppler.
- No forward flow in the pulmonary artery. In three-vessel and three-vessel- trachea views, reverse flow in late systole is seen in the main pulmonary artery via the ductus arteriosus (Fig. 2c) while the aorta appears dilated with normal antegrade flow.
- Ventricular-coronary arterial communications (VCACs) (Fig.2d) may be seen in one-third of the cases. VCACs show bidirectional turbulent flow with high velocities (50 to 150 cm/sec).

DIFFERENTIAL DIAGNOSIS

- Pulmonary stenosis – Forward flow with high velocities across the pulmonary valve.
- Tricuspid atresia with ventricular septal defect – No flow across the tricuspid valve.
- Pulmonary atresia with ventricular septal defect—the RV shows normal wall thickness. VSD is present. Major aorto-pulmonary collaterals may be seen.

ADDITIONAL POINTS

- Poor prognostic findings in Pulmonary atresia with intact ventricular septum include
  - Severe tricuspid regurgitation
  - Small tricuspid valve annulus (Z-score < −3 or - 4)
  - Small RV/LV length or width (<0.5)
  - Presence of ventricular-coronary arterial communications
  - Associated extracardiac abnormalities
  - Associated chromosomal abnormalities

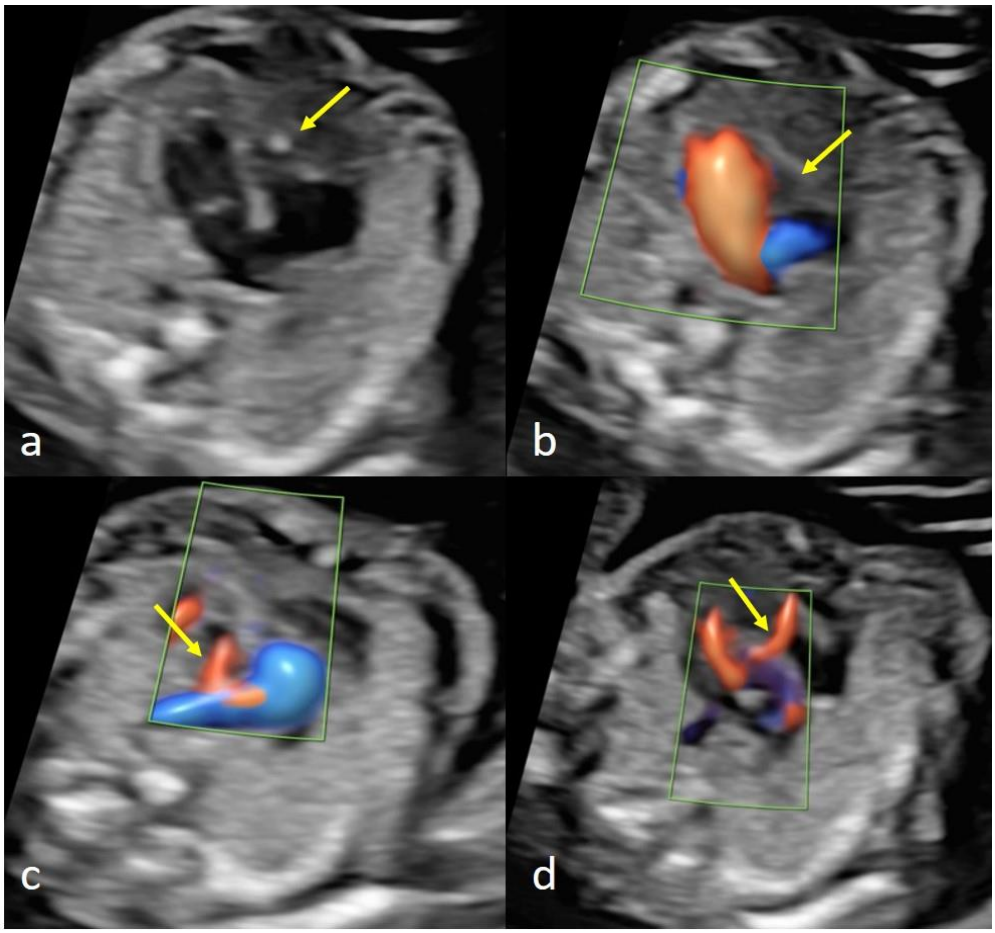


Fig.2: PAIVS (a) Four-chamber view shows a small, thick-walled RV (arrow) (b) No RV filling on color Doppler (arrow) (c) Three-vessel-trachea view shows retrograde flow in a small caliber ductus arteriosus and pulmonary artery (arrow)(d) Ventricular-coronary arterial communications (VCACs) (arrow)

DUCTUS ARTERIOSUS CONSTRICTION

Rarely, during the prenatal period, the typically patent fetal ductus arteriosus might become considerably narrow. Most cases are seen in the third trimester. In extreme circumstances, this could result in right heart failure, hydrops fetalis, and possibly death. Non-steroidal anti-inflammatory drug (NSAID) use during pregnancy is the most frequently acknowledged cause; however, many cases remain unexplained (idiopathic).

ULTRASOUND FEATURES

- Four chamber view may appear normal or may show hypertrophy of the right ventricular walls. In severe cases, the right ventricle may show poor contractility.
- Tricuspid regurgitation and pulmonary regurgitation may be present.
- Three-vessel-trachea view on color Doppler shows aliasing of flow in the ductus arteriosus near the isthmus (Fig.1, 2&3).
- Peak systolic velocity (PSV) > 140 cm /sec and end diastolic velocity (EDV) > 30 cm /sec (Figs. 2 and 3) and a pulsatility index (PI) of < 1.9 are considered diagnostic.

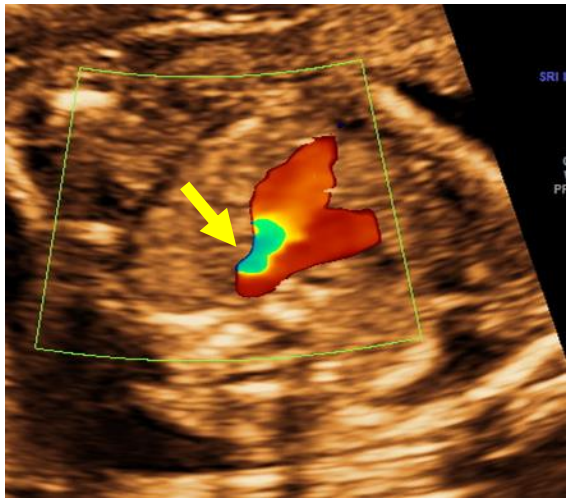


Fig. 1: Three-vessel-trachea view on color Doppler showing aliasing in the ductus arteriosus (arrow).

DIFFERENTIAL DIAGNOSIS

- Pulmonary valvular stenosis - Thickened pulmonary valve with elevated flow velocities across the pulmonary annulus.
- Fetal congestive cardiac failure – Cardiomegaly, tricuspid regurgitation and pericardial effusion. Normal flows across the ductus arteriosus.

ADDITIONAL POINTS

- Discontinuation of potential extrinsic agents, including drugs (e.g., Indomethacin and other NSAIDS) and food items containing anti-oxidants (e.g., herbal tea, coffee, chocolate, berries, etc.) results in improvement in most cases.
- Severe cases may show signs of right heart failure, which include a dilated right ventricle, pleural effusion, ascites, and body wall edema.
- Doppler of the utero-placental circulation often remains normal.

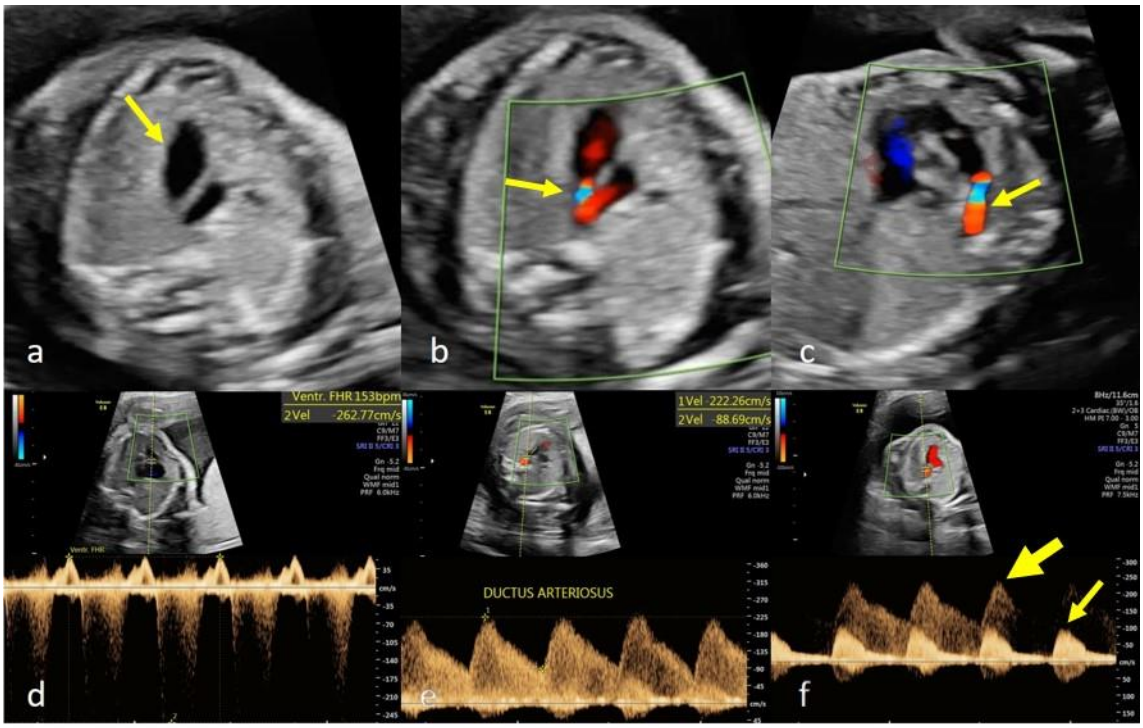


Fig. 2: Ductus arteriosus constriction in a 28 weeks fetus (a) Three-vessel-trachea view shows a dilated ductus arteriosus (arrow) with narrowing near the isthmus. (b &c) Color Doppler in three-vessel-trachea view and ductal arch view shows aliasing in the ductus arteriosus (arrows). (d) Severe pansystolic tricuspid regurgitation (e) Increased PSV and EDV in ductus arteriosus (f) Overlapping waveforms of the ductus arteriosus (large arrow) and Aortic isthmus (small arrow) show significantly elevated flow velocities in the ductus arteriosus

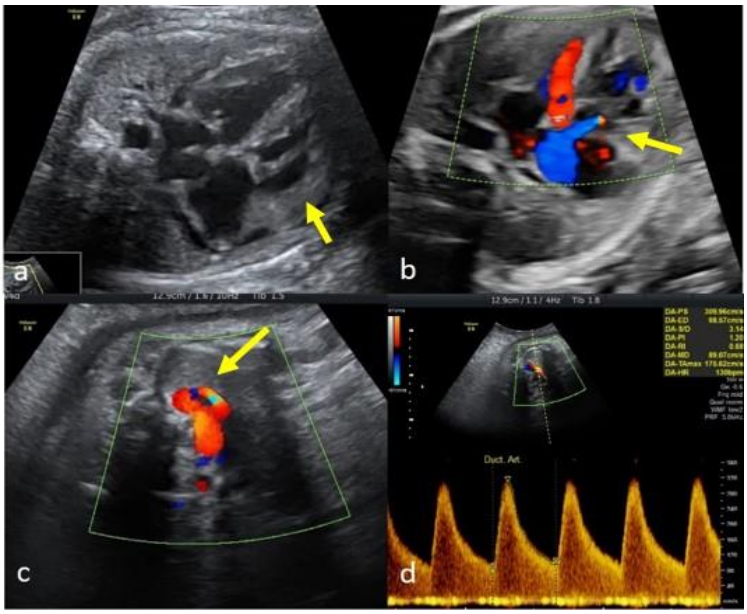


Fig. 3: Ductus arteriosus constriction in a 37 weeks fetus (a) Four-chamber view shows thickened right ventricular walls (arrow) (b) Tricuspid regurgitation (arrow). (c) Three-vessel-trachea view shows narrowing of the ductus arteriosus with aliasing(arrow) (d) Increased PSV and EDV in the ductus

VSD
WITH AN OVERRIDING VESSEL

TETRALOGY OF FALLOT

Tetralogy of Fallot (TOF) is a common cardiac defect. It is characterized by four main anatomic features:

1. Subaortic (malaligned) ventricular septal defect (VSD)
2. Overriding dilated aortic root
3. Varying degrees of right ventricular outflow tract obstruction (infundibular and valvular pulmonary stenosis)
4. Right ventricular hypertrophy is typically not present prenatally and develops postnatally owing to prolonged exposure of the right ventricular to systemic-level pressures.

ULTRASOUND FEATURES

- **Four Chamber View:** essentially normal. In some cases, a VSD may be seen, if large. In a few cases, cardiac left axis deviation is present, which may be the first clue (Fig.1 a).
- **Left Ventricular Outflow Tract (LVOT) view:** always abnormal due to a perimembranous subaortic VSD with an overriding aorta. On color Doppler, the blood streams from both ventricles into the aortic root (Y-sign). There is a loss of angle between the interventricular septum and the aortic septum. Dilated aortic root (Figs.2a & 2b)
- **Right Ventricular Outflow Tract (RVOT):** the pulmonary artery arises from the right ventricle, appears narrow but patent, and shows forward flow. The caliber of the pulmonary artery can range from almost normal-appearing to severely narrow.
- **Three Vessel Tracheal View (3VT):** abnormal due to a dilated aorta and narrow pulmonary artery. The narrow ductus arteriosus joins the aortic arch at an angle (Y-sign) instead of joining at the apex of the V. A right aortic arch is seen in 25% of cases. Color Doppler shows forward antegrade flow in cases with mild pulmonary stenosis (pink TOF). A retrograde flow suggests severe pulmonary stenosis or pulmonary atresia, resulting in duct-dependent circulation (blue TOF).

DIFFERENTIAL DIAGNOSIS

- Pulmonary atresia with VSD: pulmonary artery is very narrow and shows retrograde flow. Often accompanied by major aorto-pulmonary collaterals.
- Absent pulmonary valve syndrome (APVS): dilated pulmonary trunk and its branches with severe regurgitation from the dysplastic pulmonary valve.
- Common arterial trunk: the pulmonary artery arises not from the right ventricle but from the common trunk.

ADDITIONAL POINTS

- Aortic root dilatation is more pronounced in the third trimester.
- There is a strong association between high nuchal translucency and TOF.
- Commonly associated cardiac anomalies include a right-sided aortic arch and a persistent left superior vena cava.
- Extracardiac anomalies are also common. If multiple extracardiac anomalies are seen in association with TOF, this may be indicative of a syndromic association.
- Chromosomal abnormalities are seen in around 30% of the fetuses with TOF, and microdeletion of 22q11.2 is found in 10% to 15%.

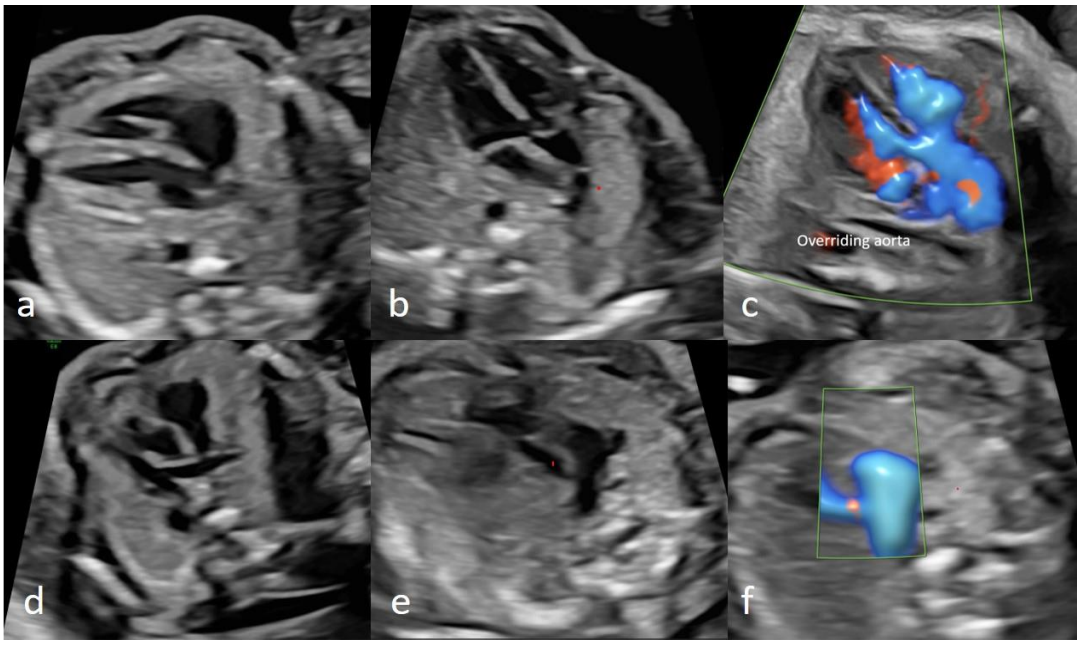


Fig. 1 (a) Four-chamber view appears normal, except for left axis deviation (b) LVOT view showing a perimembranous VSD, overriding aorta and septo aortic discontinuity (c) Color Doppler showing overriding aorta receiving blood from both the ventricles (Y-sign) (d) Right ventricular outflow tract view showing narrow pulmonary artery. (e) Three-vessel-trachea view showing narrow pulmonary artery compared to aorta (f) narrow ductus arteriosus joining the aortic arch in a branch-like fashion (Y-sign).

- The fetuses with TOF should be monitored for poor prognostic signs, which include reduced growth of the pulmonary artery, accelerated growth of the ascending aorta, cessation of forward flow through the pulmonary valve, and reversed flow through the ductus arteriosus.
- Classic TOF with pulmonary stenosis has a good prognosis, as postnatally the defect can usually be repaired in single surgery, with mortality of less than 5% during the first year of life and a good long-term outcome. However, complex cases of TOF with pulmonary atresia or absent pulmonary valve syndrome requiring more than one intervention have a less favorable outcome.

ABSENT PULMONARY VALVE SYNDROME

Absent pulmonary valve syndrome (APVS) is a rare cardiac anomaly with absent, dysplastic, or rudimentary pulmonary valve leaflets.

ULTRASOUND FEATURES

- Left axis deviation is usually present.
- Four chamber view is usually normal.
- Five chamber view shows a malaligned ventricular septal defect (VSD) with overriding aorta.
- Massively dilated main, right & left pulmonary arteries are seen in three-vessel/ three-vessel-trachea view (Bow-tie appearance) (Fig. 1,2 & 3).
- Color Doppler shows bidirectional (to-and-fro) high velocity flows across the pulmonary annulus due to severe valvular insufficiency (Fig. 2).

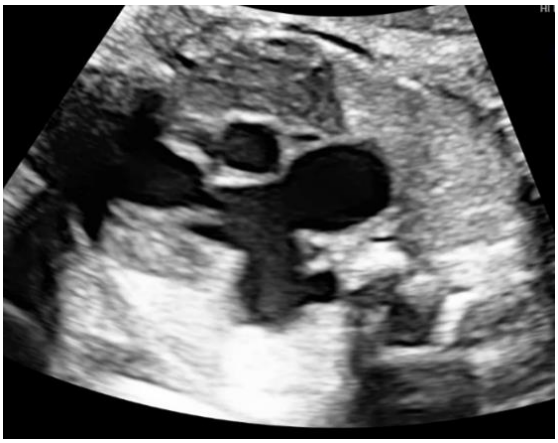


Fig.1: Three -vessel view showing grossly dilated main, right and left pulmonary arteries in APVS

DIFFERENTIAL DIAGNOSIS

- Tetralogy of Fallot (TOF)- Patent, narrow pulmonary artery with antegrade flow.
- Pulmonary atresia with VSD: Very narrow pulmonary artery with no forward flow.
- Common arterial trunk : The pulmonary artery arises from the overriding vessel.
- Double Outlet Right Ventricle: The pulmonary artery and aorta both arise from the right ventricle and course parallel to each other.

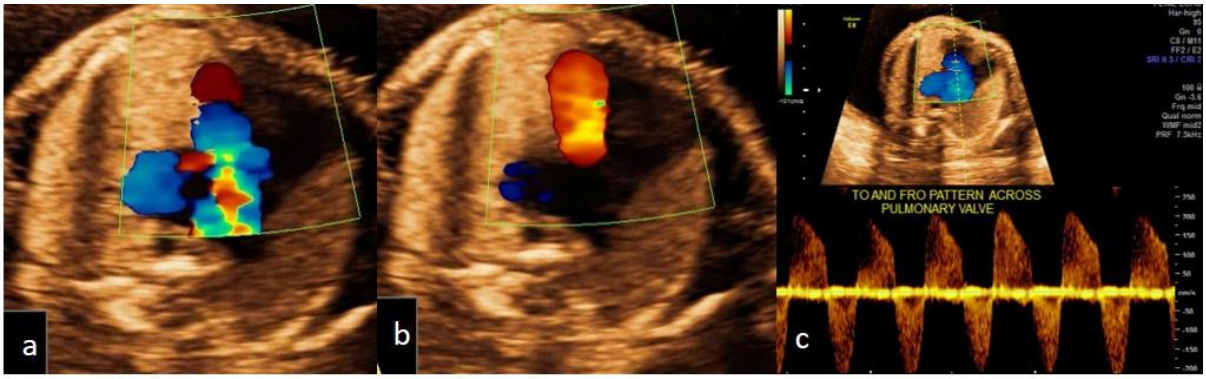


Fig.2: Color Doppler in a fetus with APVS (a) Dilated pulmonary arteries with turbulent flow across the pulmonary valve (b) severe regurgitation in diastole (c) To-an-fro flow pattern on pulse-wave Doppler.

ADDITIONAL POINTS

- Often associated with TOF & Right-sided aortic arch.
- Associated with 22q11 microdeletion in up to 20- 50 % of cases. Poor prognosis as high mortality due to cardiac failure and bronchomalacia.

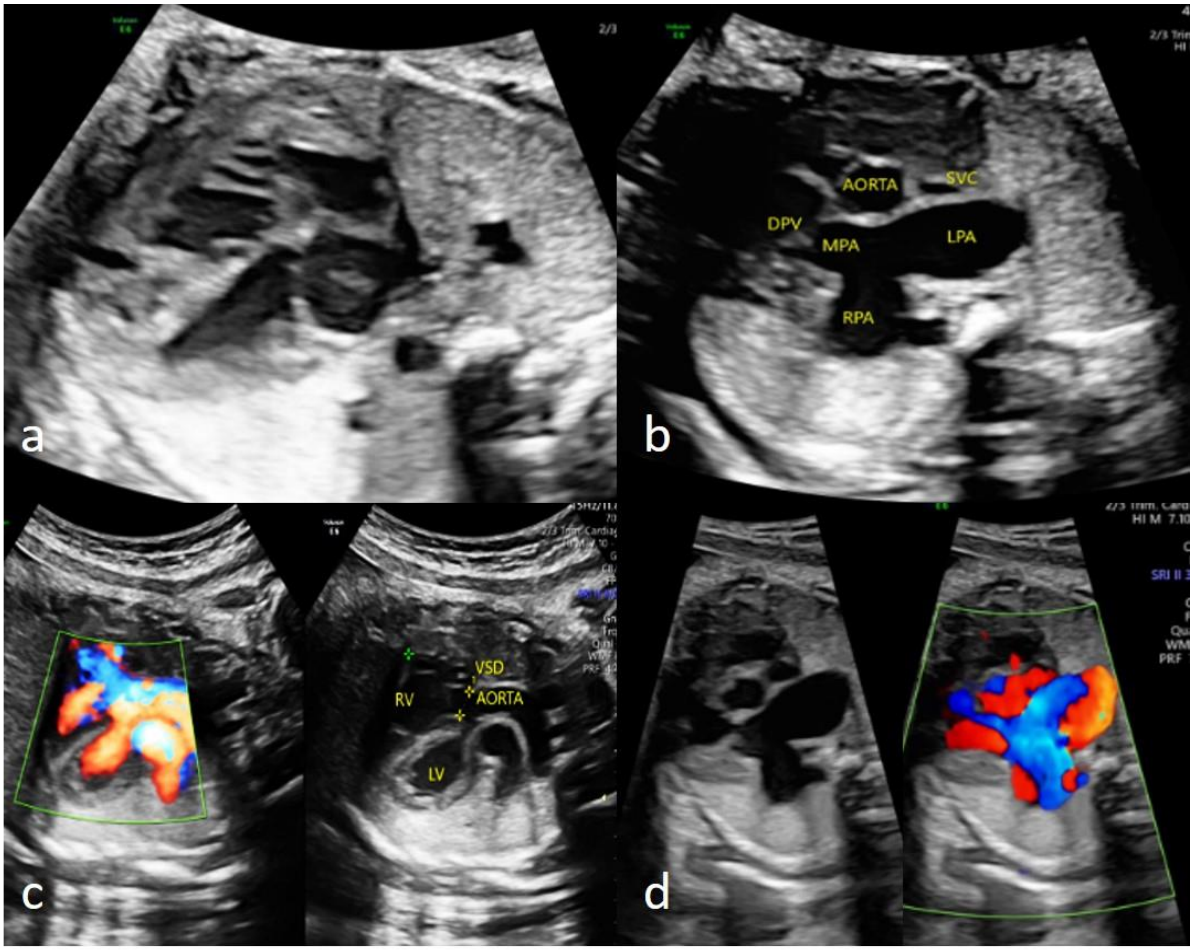


Fig.3: APVS in a 33 weeks fetus (a) Normal four- chamber view (b) Thee-vessel-trachea view showing massively dilated pulmonary artery and its branches. (c) LVOT view showing overriding of aorta (d) Color Doppler showing turbulent flow in pulmonary artery and its branches.

PULMONARY ATRESIA WITH VENTRICULAR SEPTAL DEFECT

Pulmonary atresia with ventricular septal defect (PA-VSD) is a condition characterized by atresia of the pulmonary tract with perimembranous VSD. This was previously referred to as "severe tetralogy of Fallot" (TOF).

ULTRASOUND FEATURES

- Four-chamber view is typically normal, unless associated with a large VSD (Fig. 1a).
- Left outflow tract view shows a VSD with an overriding vessel and loss of septo-aortic angle. The degree of aortic root dilatation is greater than in TOF with pulmonary stenosis, as the entire right ventricular stroke volume is directed to the aorta (Fig. 1b).
- Three-vessel view shows a dilated aorta. The pulmonary artery or ductus are difficult to visualise on gray scale.
- Color Doppler shows biventricular flow into the aorta on the LVOT view (Fig. 1c). Three-vessel view shows forward flow in the aorta and reversal of flow in the atretic pulmonary artery (Fig. 1d).
- Color doppler is also essential to demonstrate major aorto-pulmonary collaterals (MAPCAs) in the arch view (Figs. 1e and 2b).
- Ductus is seen underneath the aortic arch and is seen on the aortic arch view, below the arch (Fig. 1f).

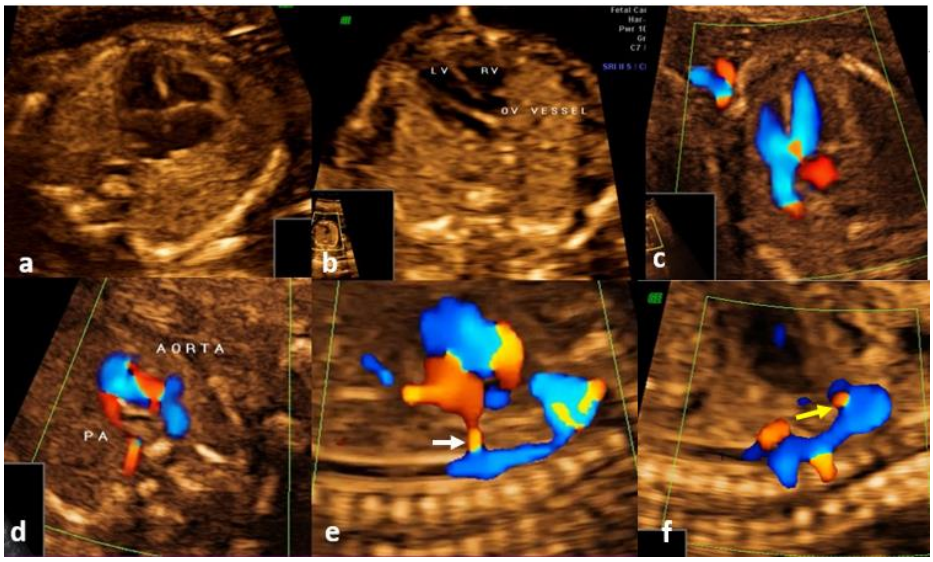


Fig. 1: PA-VSD (a) Normal four chamber view (b) LVOT view shows a perimembranous VSD with an overriding vessel (c) Color Doppler shows biventricular supply to the aorta (d) Three-vessel trachea view shows forward flow in the aorta and flow reversal in the atretic ductus (e) Arch view depicts MAPCAs (arrow). (f) Arch view depicts flow reversal in the ductus (arrow).

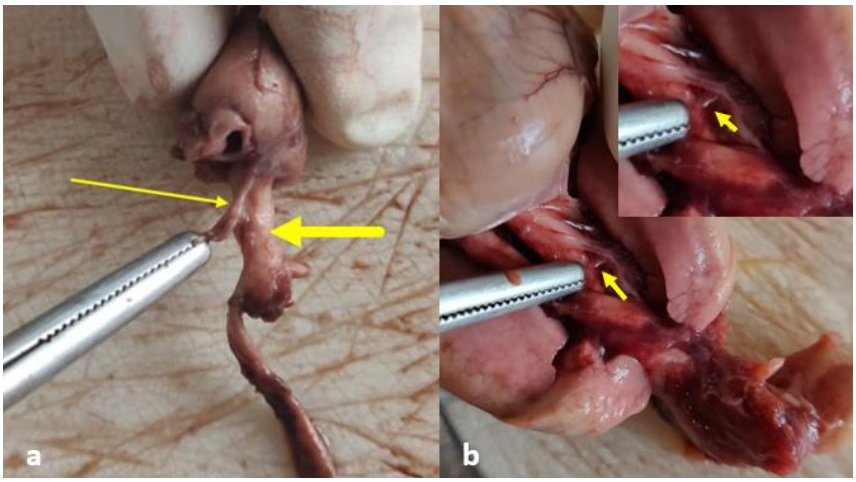


Fig. 2 (a) Autopsy specimen shows a dilated aorta (thick arrow) and an atretic pulmonary artery (thin arrow) (b) MAPCAs (arrows).

DIFFERENTIAL DIAGNOSIS

- TOF with pulmonary stenosis: the pulmonary artery and ductus are narrow but not atretic. Color Doppler shows forward flow in the pulmonary artery. MAPCAs are not visualised.
- TOF with absent pulmonary valve syndrome: markedly dilated main pulmonary artery and branch pulmonary arteries, giving a bow-tie or butterfly appearance. Color Doppler shows to-and-fro flow in the pulmonary artery. Ductus arteriosus and MAPCAs are usually absent.

ADDITIONAL POINTS

- Mothers with diabetes have a ten-fold greater risk of PA-VSD during pregnancy.
- A right-sided aortic arch is present in 20% to 50% of the cases of PA-VSD.
- Atrial septal defect (secundum type) is seen in about half the cases of PA-VSD postnatally.
- There is a high incidence of numerical chromosomal aberrations in PA-VSD, in the range of 8.3%.
- In PA-VSD, 22q11 microdeletion is found in 20% of fetuses.
- Presence of MAPCAs in PA-VSD is a risk factor for late mortality.

DOUBLE OUTLET RIGHT VENTRICLE

Double outlet right ventricle (DORV) refers to a heterogeneous group of cardiac anomalies in which the aorta and the pulmonary artery arise primarily from the right ventricle.

ULTRASOUND FEATURES

- Four-chamber view - may show a large ventricular septal defect (VSD). The left ventricle may appear smaller compared to the right ventricle (RV).
- Five-chamber view- Lack of septo-aortic continuity. Origin of both the great arteries from the anterior chamber (RV) (Figs.1 and 2).
- If one of the great vessels overrides the VSD, > 50% of it arises from the RV, and the other vessel arises completely from the RV.
- Parallel orientation of the great vessels.

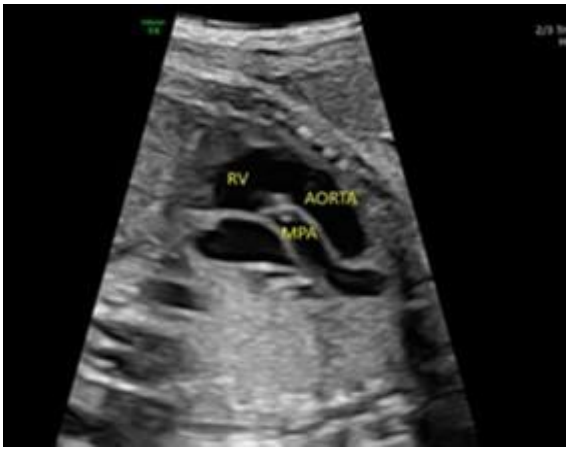


Fig. 1: Sagittal view of the fetal heart showing the aorta and the main pulmonary artery (MPA) arising from the anterior ventricle (RV) demonstrating DORV

DIFFERENTIAL DIAGNOSIS

- Tetralogy of Fallot (TOF) : the pulmonary artery arises completely from the right ventricle; the aorta overrides the VSD in such a way that it predominantly (>50%) arises from the left ventricle.
- Transposition of great arteries (TGA) : ventriculo-arterial discordance, i.e., the pulmonary artery arises from the left ventricle and the aorta from the right ventricle. In cases with a malalignment VSD, the pulmonary should predominantly (>50%) arise from the left ventricle to be classified as TGA.

ADDITIONAL POINTS

- Many types of DORV exist based on the spatial relationship of the great vessels and the location of the VSD. Three main types are **VSD type DORV**, **TOF type DORV,** and **TGA type DORV (Taussig-Bing Anomaly).** In the Taussig-Bing anomaly, the aorta arises entirely from the right ventricle, and the pulmonary artery overrides the VSD, arising predominantly from the RV (Fig. 3).
- The size disparity between the vessels, rather than Doppler flow measurements, is the best way to determine whether an obstruction is present in one of the outflows. Pulmonary stenosis is the most common associated malformation.
- Other associated cardiac malformations may include mitral atresia, atrio-ventricular septal defect, right aortic arch, coarctation of aorta, anomalous pulmonary venous connections, and persistent left superior vena cava. DORV is frequently a part of right or left isomerism.
- Extracardiac malformations are very common.
- Chromosomal abnormalities are seen in 12 to 40% of cases, primarily trisomy 18, 13, 21, and 22q11.2 deletion.

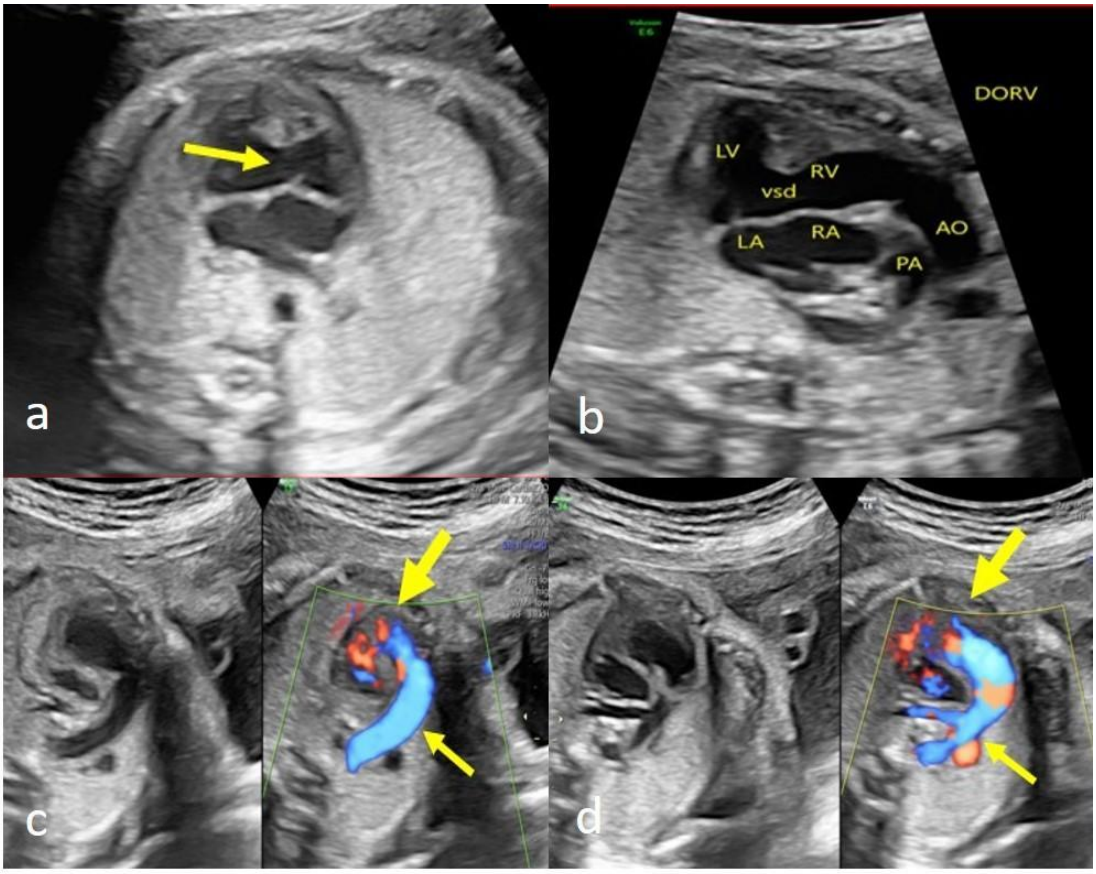


Fig 2: DORV (a) Four chamber view shows a large VSD (arrow) (b) Sagittal view showing VSD septal defect and parallel origin of great vessels arising from the RV (c) Oblique view showing a non-branching vessel (aorta) (small arrow) arising from the RV (large arrow) (d) Pulmonary artery, which is the branching vessel (small arrow) also arising from the anterior chamber (RV) (large arrow)

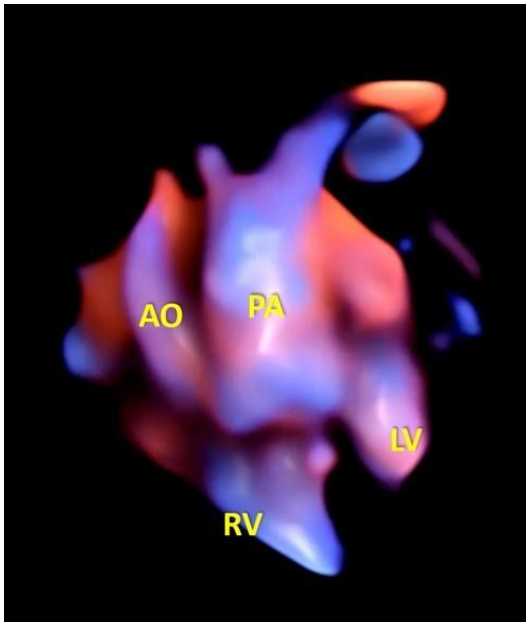


Fig 3: 3D STIC HD flow volume rendering depicting Taussig-Bing anomaly with pulmonary artery (PA) and aorta (AO) arising from the right ventricle (RV). The pulmonary artery is overriding the VSD.

COMMON ARTERIAL TRUNK

Common arterial trunk (CAT) is a condition where a single arterial trunk is seen arising from the base of the heart, and this vessel gives rise to the systemic, coronary, and pulmonary circulations. It is also referred to as “persistent truncus arteriosus” or “truncus arteriosus communis”.

ULTRASOUND FEATURES

- Four-chamber view is usually normal. Levorotation and/or a large ventricular septal defect (VSD) may be seen.
- Five-chamber view shows a malaligned VSD with an overriding large vessel.
- There is an absence of a separate pulmonary artery with a valve arising from the right ventricle. Instead, the pulmonary artery or its branches arise from the overriding vessel (Fig. 1).
- The root of the common trunk is large. The valve of the common trunk shows thickened and dysplastic leaflets, which lack proper excursion and show regurgitation on color Doppler (Fig. 2).
- Three-vessel-trachea view shows a single vessel of large caliber. However, in cases where ductus arteriosus is present, the V-sign can be seen (Fig.2 e).

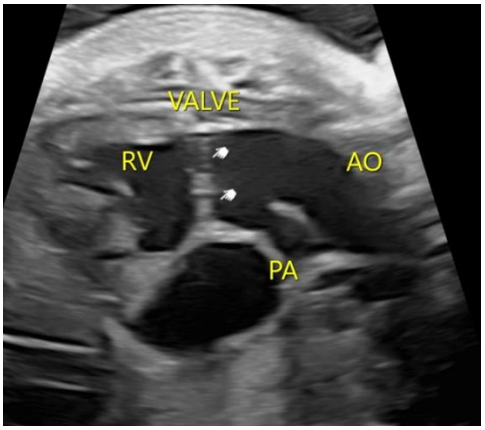


Fig. 1: The pulmonary trunk is arising from the common arterial trunk, above the level of the truncal valve

DIFFERENTIAL DIAGNOSIS

- Tetralogy of Fallot (TOF): The pulmonary artery arises from the right ventricle and shows a separate valve. The overriding vessel shows normal leaflets.
- Pulmonary atresia with ventricular septal defect (PAVSD): A severely narrow pulmonary artery arises from the right ventricle. The overriding vessel shows normal leaflets. Major aorto-pulmonary collaterals (MAPCAs) may be seen. Non-pulsatile flow in the pulmonary artery.
- Double outlet right ventricle (DORV) – The pulmonary artery and the aorta arise predominantly from the right ventricle.

ADDITIONAL POINTS

- Common arterial trunk is classified into four types based on the anatomic origin of the pulmonary artery (Collet and Edwards classification).
- Type 1: A short pulmonary trunk arises from the common trunk and divides into the right and left pulmonary arteries (Fig. 2).
- Type 2 and 3 : there is no main pulmonary artery, the right and left pulmonary arteries arise from the common trunk.
  - In Type 2 , they arise close to each other (Fig.3)
  - In Type 3, they arise at a distance from one another.

- Type 4 – The pulmonary arteries arise from the aortic arch or the descending aorta. This has been reclassified as pulmonary atresia with VSD.

- Associated anomalies include a right aortic arch, interrupted aortic arch, persistent left superior vena cava, a small or absent thymus, etc. Ductus arteriosus is absent in 50% of the cases.
- 30 to 40% cases of CAT show 22q11 microdeletion. 5% cases may show trisomy 21, 18, 13 and triploidy.

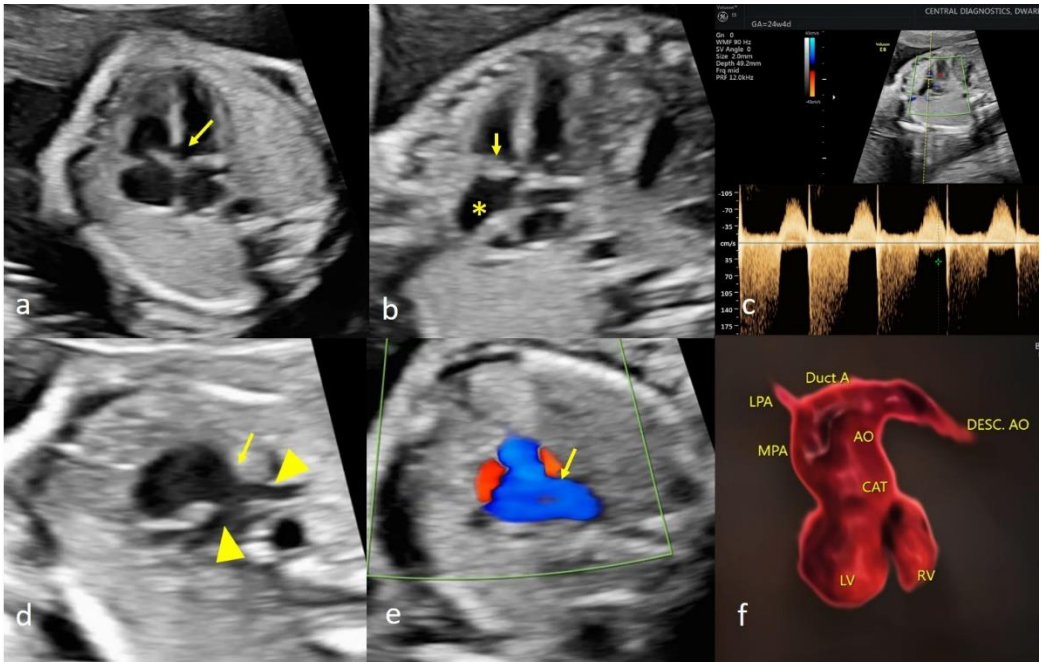


Fig. 2: CAT type 1 (a) Levorotation and VSD (b) Overriding vessel (*) with dysplastic valve (arrow) (c) Regurgitation from the dysplastic valve (d) A short pulmonary trunk (arrow) is arising from the common arterial trunk, which divides into right and left pulmonary arteries (arrowheads) (e) Ductus arteriosus was present in this case, forming a normal V sign in three-vessel-trachea view (f) 3D volume rendering showing spatial relationship of the various vessels

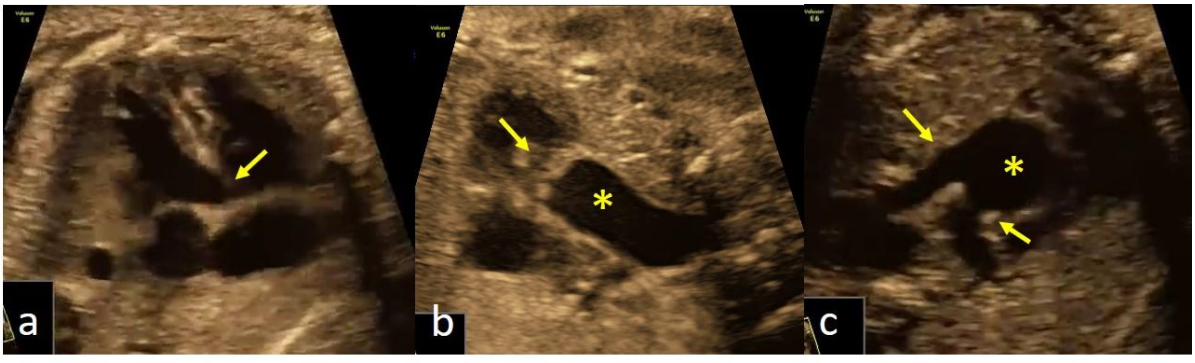


Fig. 3: CAT type 2 (a) VSD (b) Overriding vessel (*) with dysplastic valve (arrow) (c) The right and left pulmonary arteries (arrows) arise directly from the common trunk (*) close to each other. ***Case contributed by Dr Rakhi Agarwal, Patna***

TRANSPOSITION OF THE GREAT ARTERIES

TRANSPOSITION OF THE GREAT ARTERIES

Transposition of the great arteries (TGA) is a conotruncal anomaly in which the aorta arises from the morphological right ventricle and the pulmonary artery from the morphological left ventricle. This results in ventriculoarterial discordance with atrioventricular concordance. Both great arteries display a parallel course. In the majority of fetuses with TGA, the aorta is malpositioned anterior and to the right of the pulmonary artery, with a subaortic conus, and the pulmonary artery is posterior and to the left of the aorta, with fibrous continuity between the mitral and pulmonary valves. This great vessel arrangement is seen in about 88% of TGA and is referred to as d-TGA (d = “dexter”). In 12% of TGA, the spatial relations of the great arteries can be side by side.

ULTRASOUND FEATURES

- Four chamber view is typically normal (Fig.1a), except for associated ventricular septal defect (VSD), if present.
- The first great artery seen above the level of the four-chamber view is the branching pulmonary artery, i.e, the great artery arising from the left ventricle is branching (Fig. 1b).
- Aorta arises from the anterior right ventricle (Fig. 1c) in an anterior and parallel course to the pulmonary artery.
- There is no normal "crossing over" of the great arteries at the origin of the great arteries, as they arise in parallel. This parallel orientation of the great arteries is best seen in the oblique plane of the foetal heart (Fig. 1 d and e).
- In most cases with d-TGA, the three-vessel trachea view shows a single large vessel (the transverse aortic arch) with a superior vena cava to its right. The aorta can assume a right convex shape **(reverse boomerang sign)** or a straight orientation **(I sign)** (Fig. 1f).
- In the **side-by-side type of d-TGA**, the three-vessel tracheal view can appear near normal, with the pulmonary artery and aorta forming a V configuration. However, this V configuration is a little different from the normal V configuration. Here, the pulmonary artery is posterior and the aorta is anterior. Moreover, the antero-posterior length of the pulmonary artery is shorter than that of the aorta (Fig. 2 b and c).

DIFFERENTIAL DIAGNOSIS

- Double Outlet Right Ventricle (DORV): both great arteries predominantly arise from the right ventricle. A VSD is always present.
- Congenitally Corrected Transposition of the Great Arteries (ccTGA): there is an atrioventricular discordance along with a ventriculo-arterial discordance.

ADDITIONAL POINTS

- VSD and pulmonary stenosis are commonly associated cardiac defects in d-TGA
- VSDs are common, occur in up to 40% of cases. Typically perimembranous, these can be located anywhere along the septum.
- Pulmonary stenosis coexists with a VSD in d-TGA patients in up to 20% of cases, and the stenosis is usually more severe and complex than in d-TGA with an intact ventricular septum.

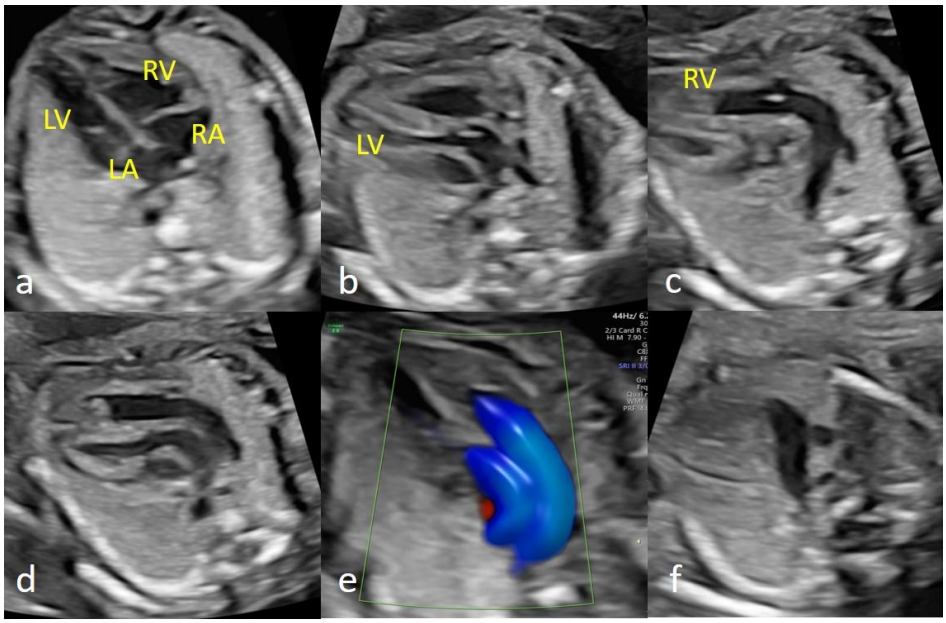


Fig. 1: Transposition of the great arteries (a) Normal four-chamber view (b) LVOT view shows the great artery arising from the left ventricle (LV) is branching (pulmonary artery) (c) The great artery arising from the right ventricle (RV) is non-branching (aorta) (d & e) Oblique views in grayscale and color showing parallel great arteries (f) Three-vessel-trachea view shows a single great artery, which is the aorta, with a straight course (I sign).

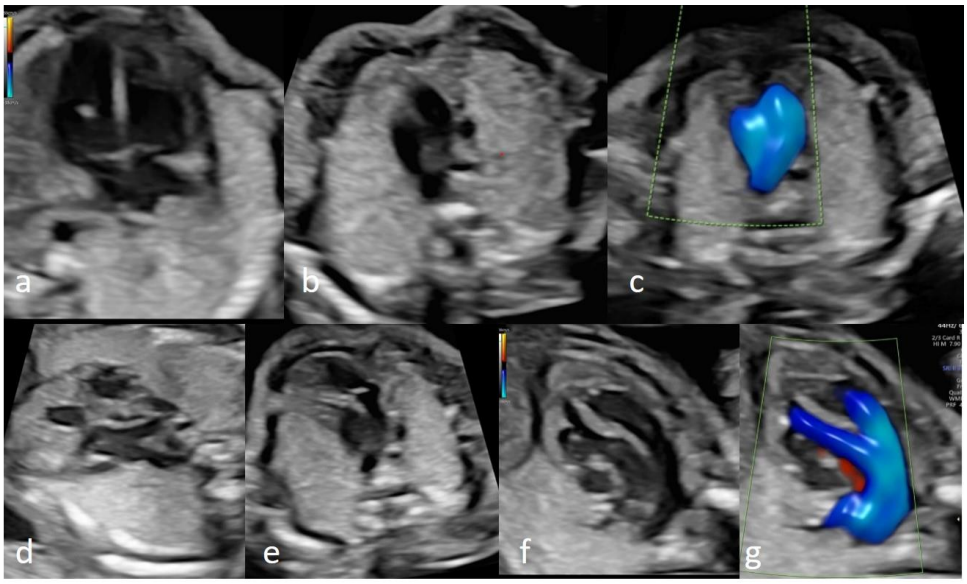


Fig.2 TGA with side-by-side arrangement of the great vessels (a) Normal four chamber view (b &c) Three-vessel-trachea view on grayscale and color Doppler shows a V sign. However, note that the pulmonary artery on the left side is posterior to the aorta and appears shorter in length than the aorta (d) Pulmonary artery (branching artery) arising from the left ventricle (e) Aorta arising from the anterior ventricle (right ventricle) (f & g) Oblique views in grayscale and color show parallel great arteries

CONGENITALLY CORRECTED TRANSPOSITION OF THE GREAT ARTERIES

Congenitally corrected transposition of the great arteries (CCTGA) is a rare cardiac abnormality characterized by atrioventricular and ventriculoarterial discordance.

ULTRASOUND FEATURES

- **Atrioventricular discordance** : the morphological right atrium (RA) is connected to the morphological left ventricle (LV) by the mitral valve and the morphological left atrium (LA) is connected to the morphological right ventricle (RV) by the tricuspid valve (Fig. 1 b and c, Fig. 2).
- **Ventriculoarterial discordance** -The pulmonary artery (PA) is connected to the morphological LV and the aorta (AO) to the RV (Fig.1 d & e, Fig.2)
- **Levo-transposition of the great vessels** (L-TGA)- Aorta is located anterior and to the left of the pulmonary (Fig. 1e)
- RV is left-posterior, identified by the moderator band. LV is right-anterior, having a smooth surface and an elongated appearance.

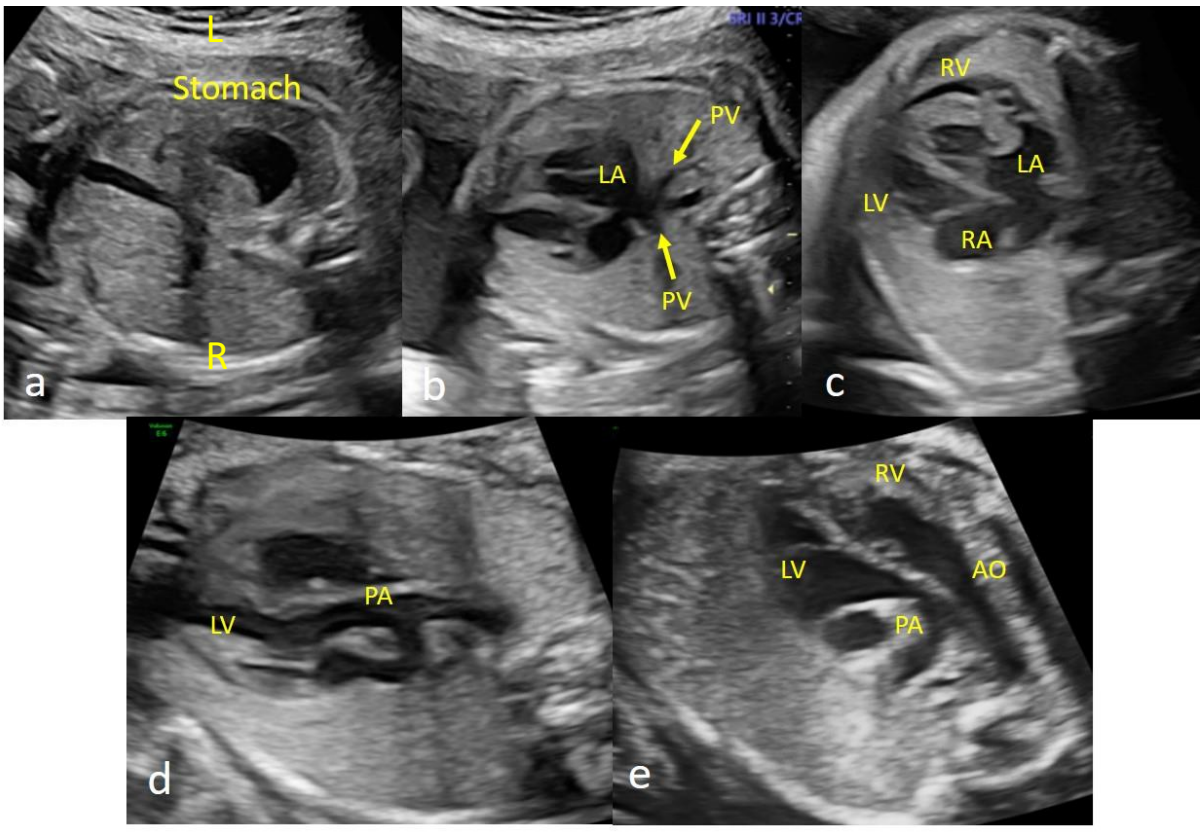


Fig. 1: CCTGA. Fetus in breech presentation (a) Situs solitus. Stomach on the left side (b) Cardiac apex along the midline (mesocardia). Pulmonary veins (PV) draining into the left atrium (LA)(c) Atrioventricular discordance. LA is connected to a thick-walled morphological right ventricle (RV) with a moderator band, and right atrium (RA) is connected to the morphological left ventricle (LV) (d) Ventriculoarterial discordance- the pulmonary artery (PA, the branching great vessel) arising from the morphological left ventricle (e) The aorta (AO) arising from the morphological right ventricle. Parallel arrangement of the great vessels.

DIFFERENTIAL DIAGNOSIS

- DORV: both great vessels predominantly arise from the RV.
- D-TGA: atrioventricular concordance is maintained. The pulmonary artery arises from the left sided ventricle (pulmonary artery in CCTGA arises from the right sided morphological left ventricle).

ADDITIONAL POINTS

- Congenitally corrected transposition of the great arteries is associated with situs inversus in 5% of cases.
- Mesocardia is seen in 25% of CCTGA patients and is often the first clue to the diagnosis.

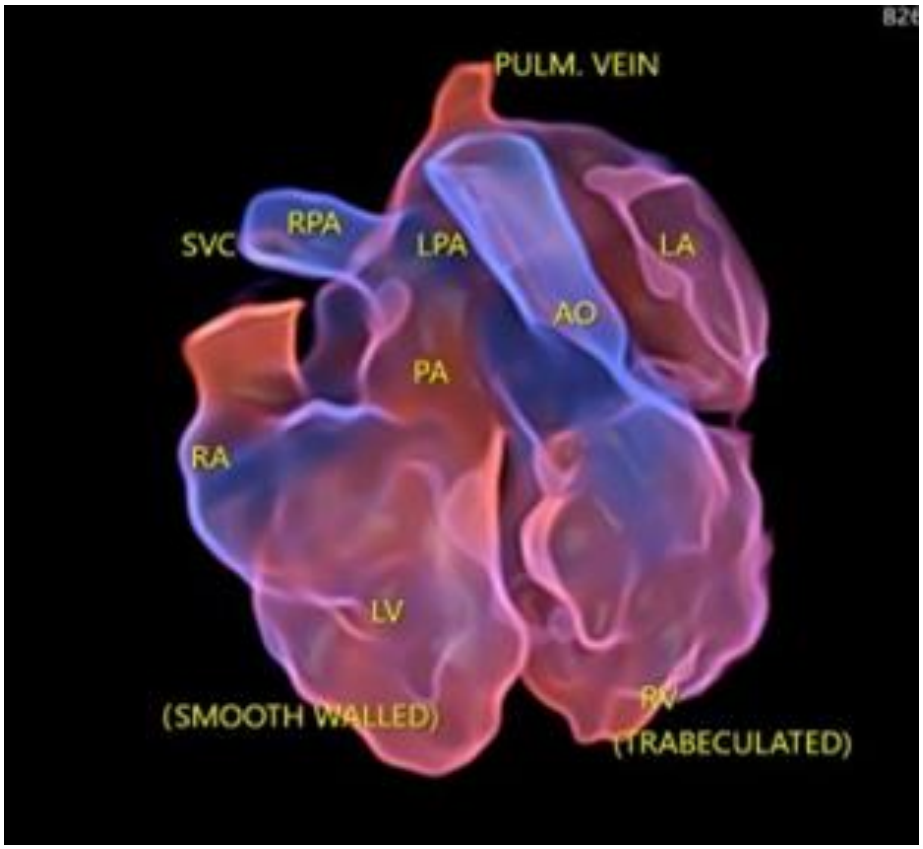


Fig. 2: STIC HD-flow volume rendering depicting the connections in CCTGA. The pulmonary veins (Pulm.vein) are draining into the left atrium (LA), which is connected to a trabeculated right ventricle (RV). Superior vena cava (SVC) is draining into the right atrium (RA), which is connected to the smooth walled left ventricle (LV). Hence, there is atrioventricular discordance. The pulmonary artery (PA) is connected to the morphological left ventricle and the aorta (AO) is arising from the morphological right ventricle (ventriculoarterial discordance)

AORTIC ARCH ANOMALIES

COARCTATION OF AORTA

Coarctation of aorta (CoA) is characterized by the narrowing of the aortic arch, typically located in the isthmic region between the left subclavian artery and the ductus arteriosus. When the narrowing involves a long segment of the aortic arch, it is called **tubular hypoplasia of the aortic arch**. CoA can be classified as **simple** when it occurs without important intracardiac abnormalities and **complex** when it occurs in association with significant intracardiac abnormalities.

ULTRASOUND FEATURES

- Four-chamber view: Ventricular disproportion. Left ventricle (LV) smaller than right ventricle (RV) (Fig. 1a) is the most common finding that raises the suspicion for Coarctation.
- The ratio of right ventricular and left ventricular width, reported as 1.19 in normal fetuses, is $\geq$1.69 in fetuses with coarctation of the aorta.
- Three-vessel view: Small to normal calibre of the ascending aorta (Fig. 1b).
- Three-vessel-trachea view: reduced calibre of the aortic isthmus (Fig. 1c). Isthmus: duct ratio < 0.74
- Ratio of the left to right sided structures (LV/RV, Mitral valve annulus /Tricuspid valve annulus diameter, Aortic Valve annulus /Pulmonary valve annulus diameter) <0.6 or Pulmonary artery (PA) /Aorta (AO) ratio >1.6. Great artery ratio has the highest sensitivity.
- Transverse aortic arch diameter < 3mm has the highest sensitivity for diagnosis.
- Aortic arch view (longitudinal plane): The aortic arch appears narrow (Fig. 1d) and often elongated between the left subclavian artery origin and the isthmus. There is a shelf like narrowing along the posterior-lateral wall of the aorta at the isthmus **(Contraductal shelf).**
- Aortic arch view: Aortic ductal angle- 82.2-125° (Normal 128.2-167°)
- Color Doppler: Forward flow across the aortic valve. Mostly forward flows across the aortic arch narrowing with no increase in velocities. Bidirectional or reversal of flow in aortic isthmus in severe cases.
- Persistent left superior vena cava may be seen occasionally.

DIFFERENTIAL DIAGNOSIS

- Severe aortic stenosis or atresia: Incomplete opening of the aortic valve with turbulent aliasing flow across the aortic valve and reversal of flow.
- Interrupted aortic arch: Non visualisation of the transverse aortic arch with a straight upward course of the aorta towards the neck.

ADDITIONAL POINTS

- Coarctation of Aorta is common, accounts for 8% of the cardiac defects.
- False positive and false negative diagnosis are very common.
- Can be associated with persistent left superior vena cava, bicuspid aortic valve, mitral stenosis, aneurysm of the foramen ovale, and chromosomal aberrations like Turner's syndrome.

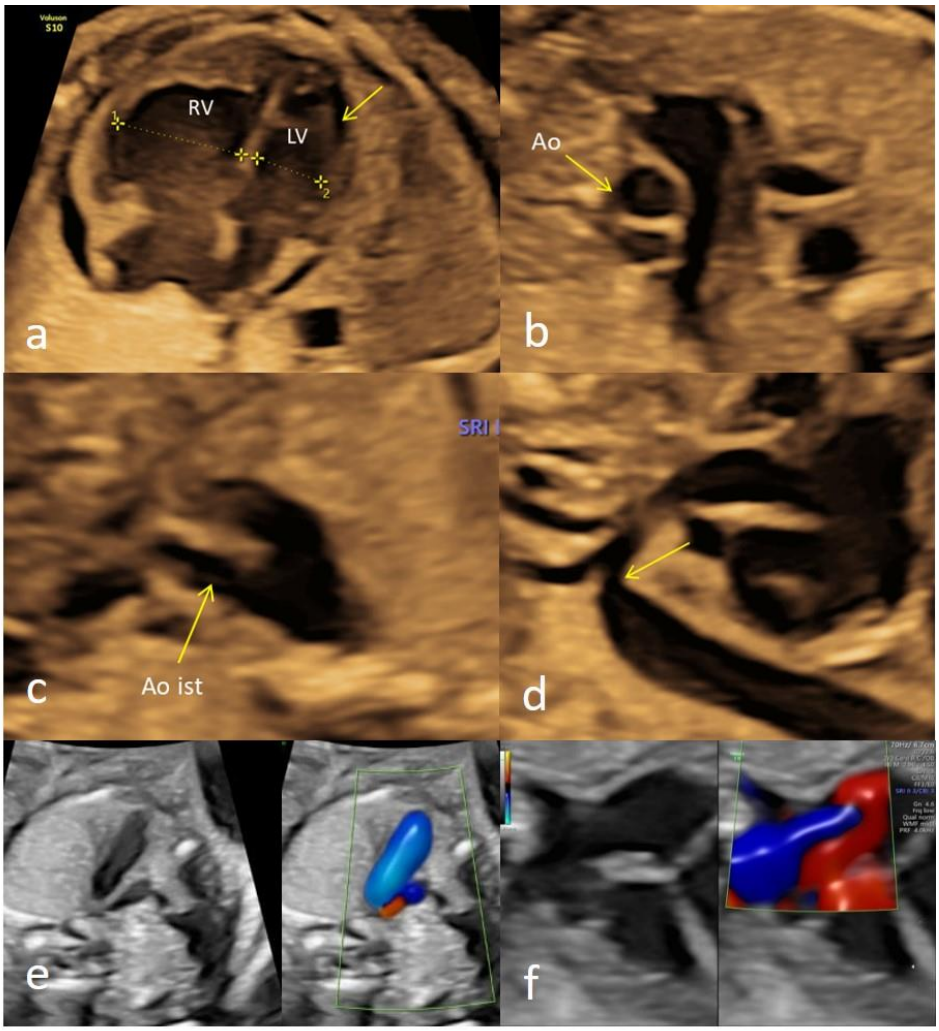


Fig. 1: Coarctation of Aorta (CoA) (a) Four chamber view : small left ventricle (b)Three-vessel view: narrow calibre of the aorta (c) Three-vessel-trachea view: narrow aortic isthmus (isthmus: duct ratio- 0.72) (d) Aortic arch view: focal narrowing of the aortic arch distal to the origin of the left subclavian artery (e) Three-vessel-trachea view in grayscale and color Doppler: narrow aortic arch with reversal of flow (f) Four-chamber view: bidirectional flow across the foramen ovale

Case contributed by Dr Sri Sai Lakshmi B J, Hyderabad

INTERRUPTED AORTIC ARCH

Interrupted aortic arch (IAA) is a malformation characterized by complete separation of the ascending and descending aorta. IAA is classified as type A, B, or C based on the anatomic location of the interruption in relation to the brachiocephalic vessels.

- Type A: The arch is interrupted after the origin of the left subclavian artery (LSA).
- Type B: Interruption between the origins of the left common carotid artery (LCA) and the LSA.
- Type C: Interruption between the brachiocephalic artery (BA) and the LCA.

Type B is the most common and is noted in about 50% to 75% of IAA cases. It is also associated with a large malalignment-type ventricular septal defect (VSD), with posterior displacement of the infundibular septum in 90% of cases. IAA type A has similar hemodynamic characteristics as coarctation of the aorta; type C is very rare.

ULTRASOUND FEATURES

- Four-chamber view shows ventricular disproportion with a small sized left ventricle (LV) in Type A IAA (Fig. 1). Four-chamber view is generally normal in Type B IAA due to a VSD, but occasionally a small sized left ventricle can be noted (Fig. 2).
- LVOT view may show VSD and a narrow aortic root.
- Three-vessel trachea view shows:
  - Lack of continuity of the aortic arch (Figs. 1b, 2b, and c).
  - Trachea appears in close proximity or touching the pulmonary artery or ductus, because of the absence of the medially located aortic arch.
  - Dilated pulmonary trunk.
  - Thymus may be absent or hypoplastic when associated with 22q11.2 microdeletions.
  - Anterior displacement of the pulmonary artery in proximity to the sternum may be a clue for thymic aplasia/hypoplasia.
- Longitudinal view of the aortic arch fails to show the typical “candy-cane” curvature. Rather, a straight course of the aorta towards the neck is seen (Figs. 1c & 2d). The aorta shows two branches (BA and LCA) in type B IAA (V pattern) (Fig. 2d) and three branches (BA, LCA and LSA) in type A IAA (W pattern) (Fig. 1d).

ADDITIONAL POINTS

- Associated cardiac defects include right aortic arch, aberrant right or left subclavian artery, atrioventricular septal defect, single ventricle, double outlet right ventricle, etc.
- About 50% of the cases of Type B IAA are associated with 22q11.2 microdeletion.

DIFFERENTIAL DIAGNOSIS

- Coarctation of Aorta or Tubular Arch Hypoplasia with VSD: in both cases, the aortic arch maintains its curvature, whereas in IAA, the aorta takes a straight course from the left ventricle into the upper chest and neck.

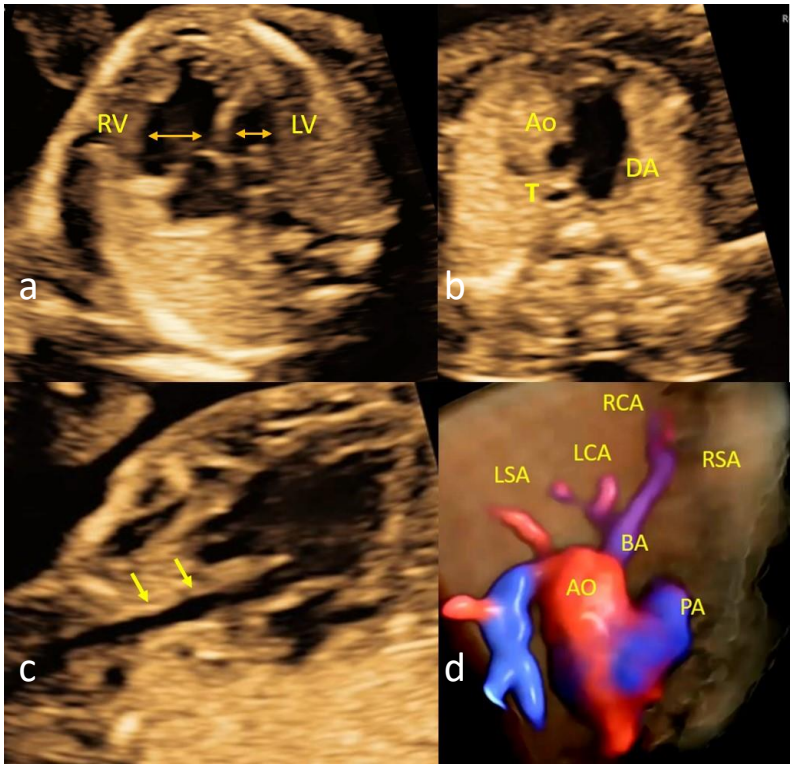


Fig. 1: Interrupted aortic arch Type A (a) Four-chamber view: ventricular disproportion (LV smaller than RV) (b) Three-vessel-trachea view in grayscale: absence of the continuity of the aortic arch (Ao) with descending aorta. Trachea (T) is in close proximity to the Ductal arch (DA) (c) Sagittal view: straight course of the ascending aorta into the neck (arrows) (d) STIC color flow volume rendering depicting the ascending aorta (AO) branching into three vessels.

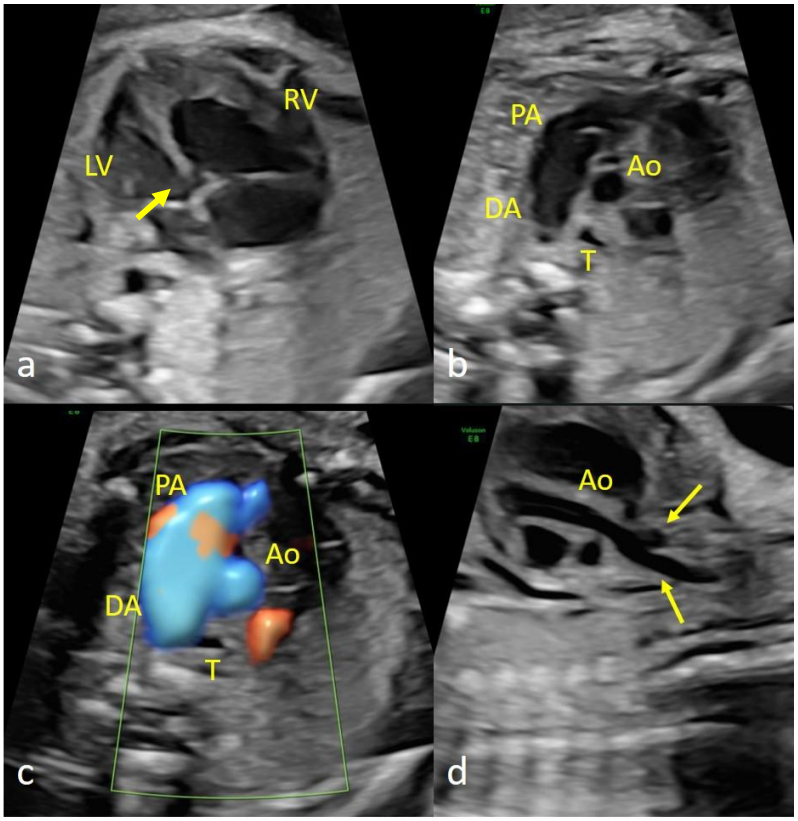


Fig.2: Interrupted aortic arch Type B (a) Four-chamber view: ventricular disproportion (LV smaller than RV) & VSD (arrow) (b and c) Three-vessel-trachea views in grayscale and with color Doppler show the aortic arch not joining the ductal arch. There is an absence of continuity of the aortic arch and the descending aorta. Trachea is in close proximity to the ductal arch (d) Sagittal view shows a straight course of the ascending aorta into the neck, branching into two vessels (arrows).

RIGHT AORTIC ARCH WITH LEFT DUCTUS ARTERIOSUS

An aortic arch that crosses across the right bronchus is known as a “right aortic arch” (RAA), meaning that the transverse aortic arch is situated on the right side of the trachea rather than the left. In the most common arrangement, the ductus arteriosus and pulmonary artery are situated to the left of the trachea, and the ductus joins the right aortic arch behind the trachea in a U-shape. This forms a loose vascular ring around the trachea. This configuration is called the right aortic arch with left ductus arteriosus (RAA-LDA).

ULTRASOUND FEATURES

- Four-chamber view shows normal cardiac chambers. The descending aorta shows a central position anterior to the fetal spine (Fig. 1a).
- Three-vessel and three-vessel-trachea views show the ascending aorta and the transverse aortic arch to the right of the trachea.
- In the same views, the pulmonary artery and the ductus are seen coursing to the left of the trachea.
- A ‘U’ shaped configuration is seen in three-vessel-trachea view, instead of a normal V **(U-sign)** as the ductus and the right aortic arch join behind the trachea. This can be seen on gray-scale as well as on color Doppler (Fig.1b&c).
- Color Doppler can also show an **aberrant left subclavian artery (ALSA)** in almost all cases in an oblique view of the upper mediastinum (Fig.2). It arises behind the trachea from the region of the junction of the ductus and the descending aorta **(Kommerell diverticulum)**.

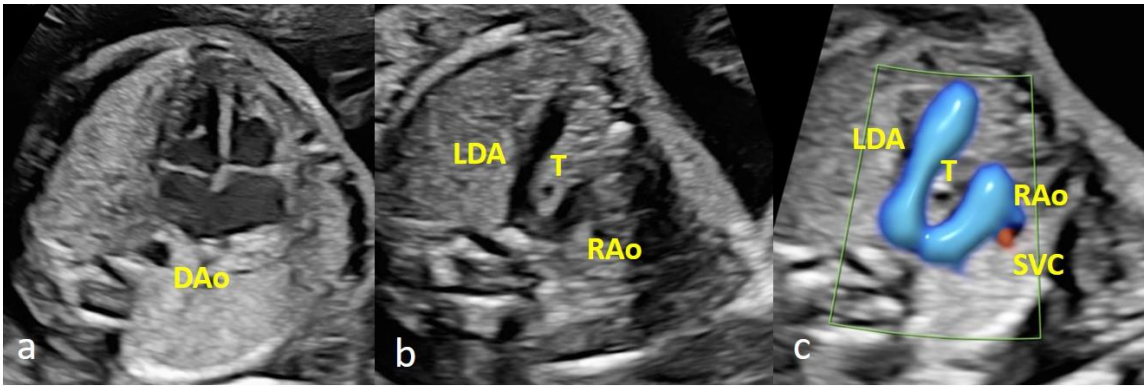


Fig. 1: RAA-LDA (a) normal four-chamber view of the heart. The descending aorta (DAo) is anterior to the spine in the central position (b & c) Three-vessel-trachea view shows a U-sign formed by the left sided ductus arteriosus (LDA) and the right aortic arch (RAo) joining posterior to the trachea (T)

DIFFERENTIAL DIAGNOSIS

- Right-sided aorta of isomerism or situs inversus: the aortic arch is to the right of the trachea but the abdominal aorta is left sided.
- Double aortic arch: the ascending aorta bifurcates into two branches, which circle the trachea. One of the limbs may be hypoplastic, thus not well seen on prenatal ultrasound. Such cases may be difficult to differentiate from RAA-LDA.

ADDITIONAL POINTS

- Associated anomalies include conotruncal anomalies (rare), persistent left superior vena cava (PLSVC) and hypoplastic or absent thymus (Fig.3).
- 22q11 microdeletion is found in 8.5% of isolated RAA cases. Hence, even in isolated cases, karyotyping or microarray should be advised.

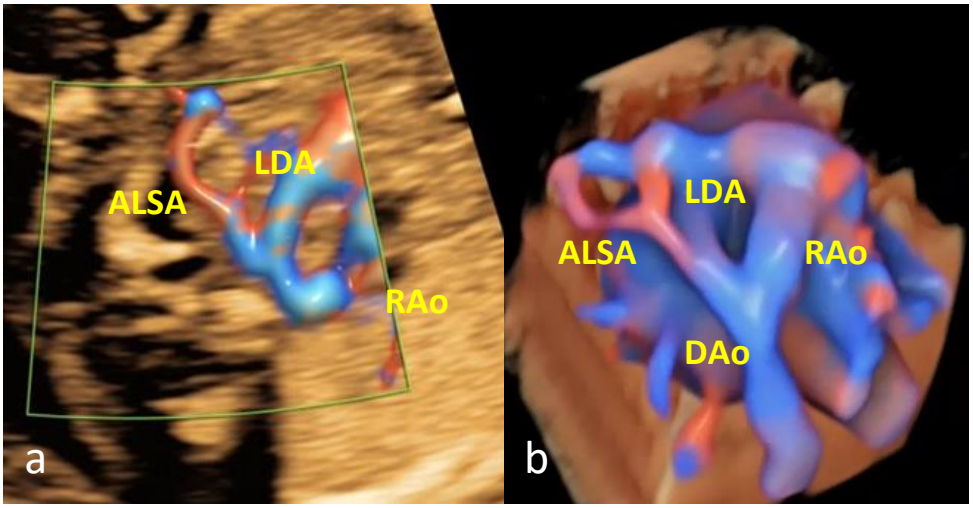


Fig. 2: Aberrant left subclavian artery (ALSA) (a) Color Doppler in an oblique view of the upper mediastinum shows ALSA arising from the junction of the left sided ductus (LDA) and right aortic arch (RAo) (b) STIC color flow volume rendering shows the spatial relationships of RAA-LDA from the posterior aspect. ALSA is arising from LDA-Descending aorta (DAo) junction.

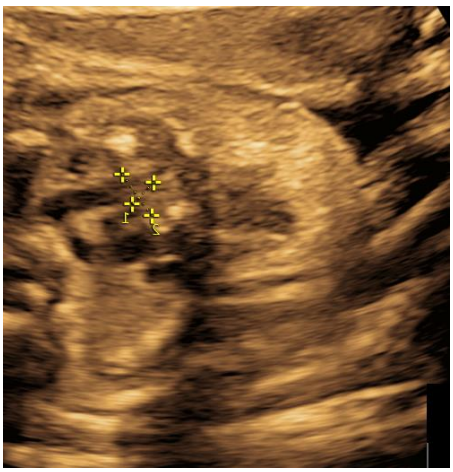


Fig. 3: RAA-LDA with hypoplastic thymus. 22q11 deletion was found on microarray

RIGHT AORTIC ARCH WITH RIGHT DUCTUS ARTERIOSUS

In this anomaly, the aortic arch and the ductus arteriosus (DA) are both on the right side of the trachea.

ULTRASOUND FEATURES

- Right V-sign: The aortic arch and the right-sided ductus join together on the right side of the trachea (Figs.1 and 3).
- Cardiac apex is towards the left.
- Most cases are associated with cardiac anomalies, usually cono-truncal malformations.
- In cases with cardiac anomalies, DA may be difficult to identify as it may be very small in caliber, tortuous, or hidden underneath the aortic arch.
- A mirror-image pattern of the aortic branching is seen (the first branch to arise from the aortic arch is the left subclavian artery, followed by the left common carotid, then the brachiocephalic trunk) (Fig. 2).

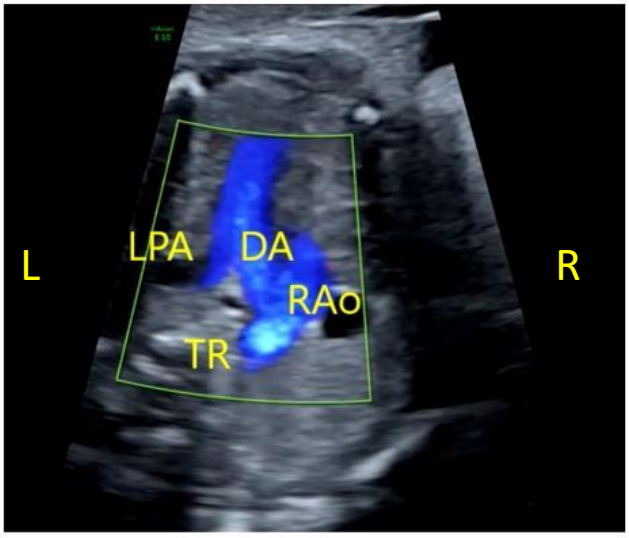


Fig. 1: Right V-sign in three -vessel trachea view, formed by Ductus arteriosus (DA) and right aortic arch (RAo) joining on the right side of the trachea (TR)

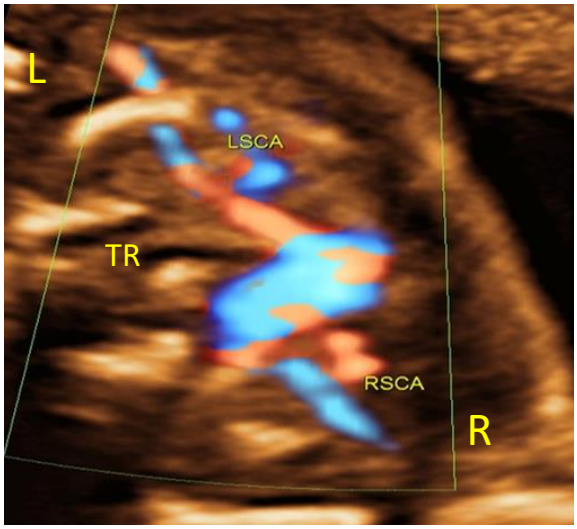


Fig. 2: Right aortic arch with mirror-image branching pattern. The first branch is the left subclavian artery (LSCA). The DA is not visible in this case.

DIFFERENTIAL DIAGNOSIS

- Situs inversus totalis: the cardiac chambers and the great vessels are oriented in a mirror-image of the normal.

ADDITIONAL POINTS

- Associated with 22q11 microdeletion

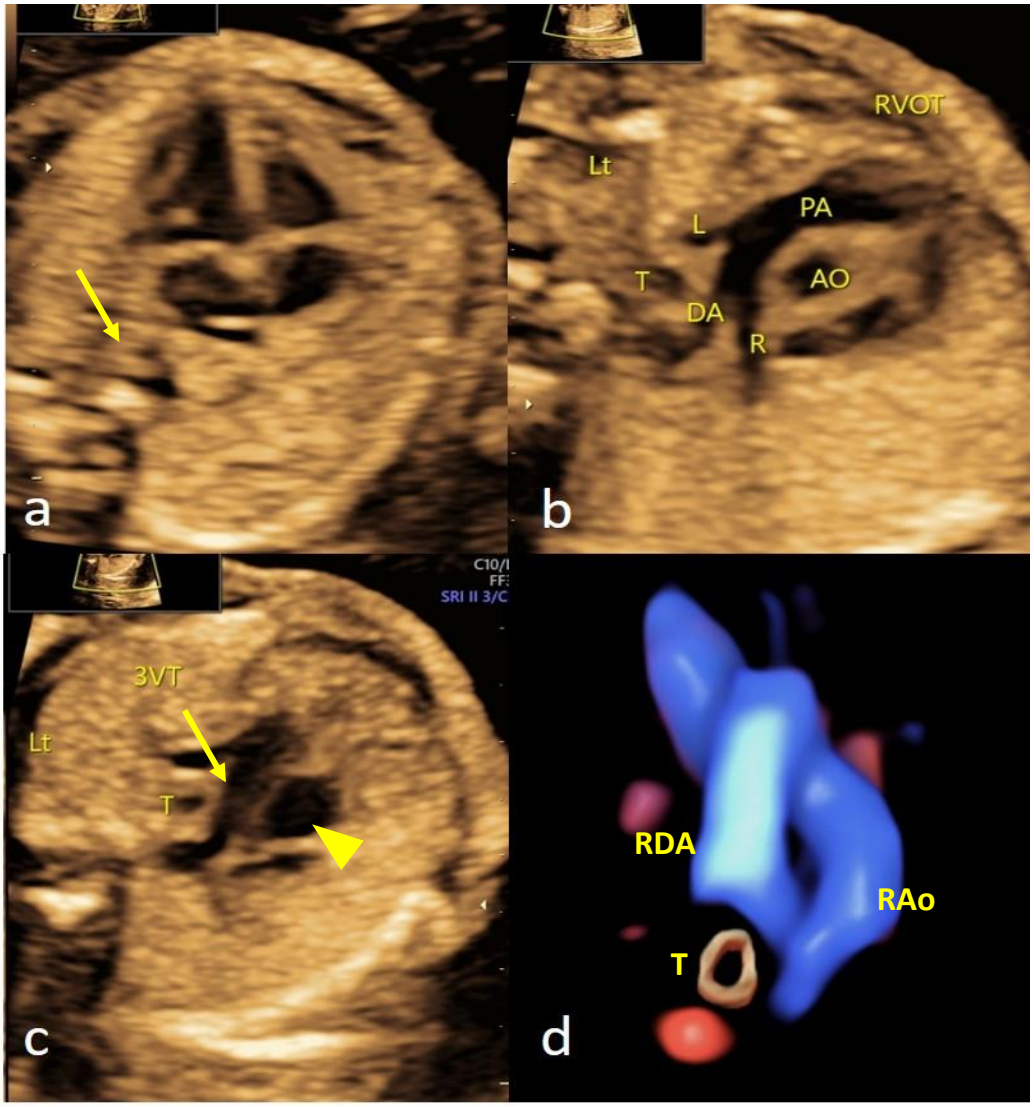


Fig 3: Right aortic arch with right ductus. (a) Normal four-chamber view. Note the centrally located descending aorta over the fetal spine (arrow) (b) RVOT view shows the ductus arteriosus (DA) crossing on the right side of the trachea (T) (c) Three-vessel-trachea view shows the aorta (arrowhead) and ductus (arrow) joining together on the right side of the trachea (T) (d) STIC color flow volume rendering showing the spatial relationships. RAo=right aortic arch, RDA= right ductus arteriosus, T= trachea ***Case contributed by Dr Alpesh D Pancholi, Vadodara***

DOUBLE AORTIC ARCH

Double aortic arch (DAA) results from the persistence of the embryonic right aortic arch (RAA) and left aortic arch (LAA). The left ductus arteriosus (LDA) persists, whereas the right ductus arteriosus regresses. Each aortic arch gives rise to a subclavian and a common carotid artery. Double aortic arch forms a tight vascular ring around the trachea and esophagus, which usually necessitates a postnatal surgical intervention.

ULTRASOUND FEATURES

- The aortic arch has a course to the right side of the trachea but bifurcates directly at the level of the trachea to have one arch to the right and one to the left (Fig. 1). This is described as a "λ" (Lambda) configuration.
- Both arches fuse into the descending aorta behind the trachea. The esophagus and the trachea are entrapped between the RAA and LAA, which form a complete vascular ring.
- The descending aorta has a course central and anterior to the spine.
- In some cases, the left arch is narrower than the right and can even be severely hypoplastic.
- The ductus arteriosus has a left sided course and connects with the left arch or the descending aorta.
- Two vessels arise from each aortic arch, i.e., a left common carotid and left subclavian artery and a right common carotid and right subclavian artery (Fig. 2).

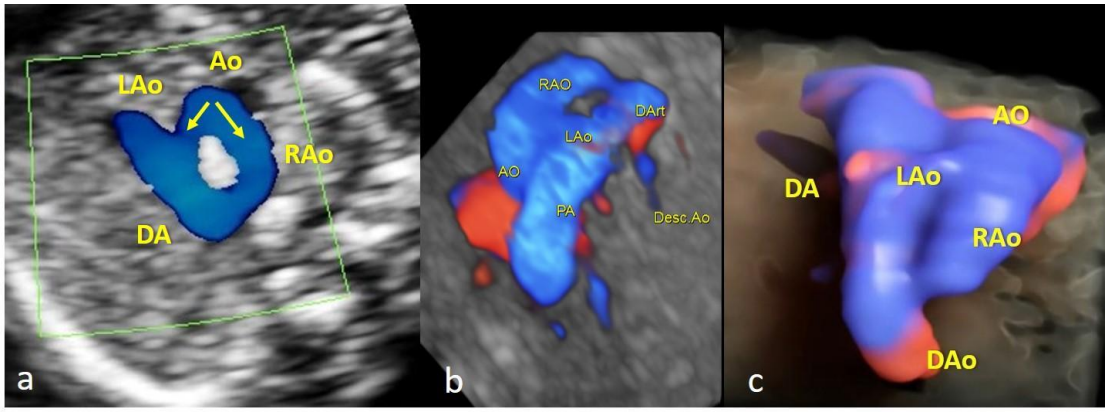


Fig. 1: Double aortic arch (a) Ascending aorta bifurcating into left (LAo) and right (RAo) aortic arches. The LAo joins the left ductus (DA). LAo, RAo and LDA form a complete vascular ring around the trachea (b & c) STIC color flow shows spatial relationships of the vessels in a Double aortic arch.

DIFFERENTIAL DIAGNOSIS

- Right Aortic arch with left ductus arteriosus (RAA-LDA): differentiating RAA-LDA from a DAA with a hypoplastic left arch can be difficult prenatally. Also, the left common carotid artery in RAA-LDA overlaps the left sided ductus; this can be mistaken as DAA.

ADDITIONAL POINTS

- DAA forms a tight vascular ring encircling both trachea and oesophagus. Compression of the trachea may cause stridor and tracheomalacia in neonates.
- 3D color STIC is a helpful additional tool for depiction of aortic arch anomalies, particularly DAA.

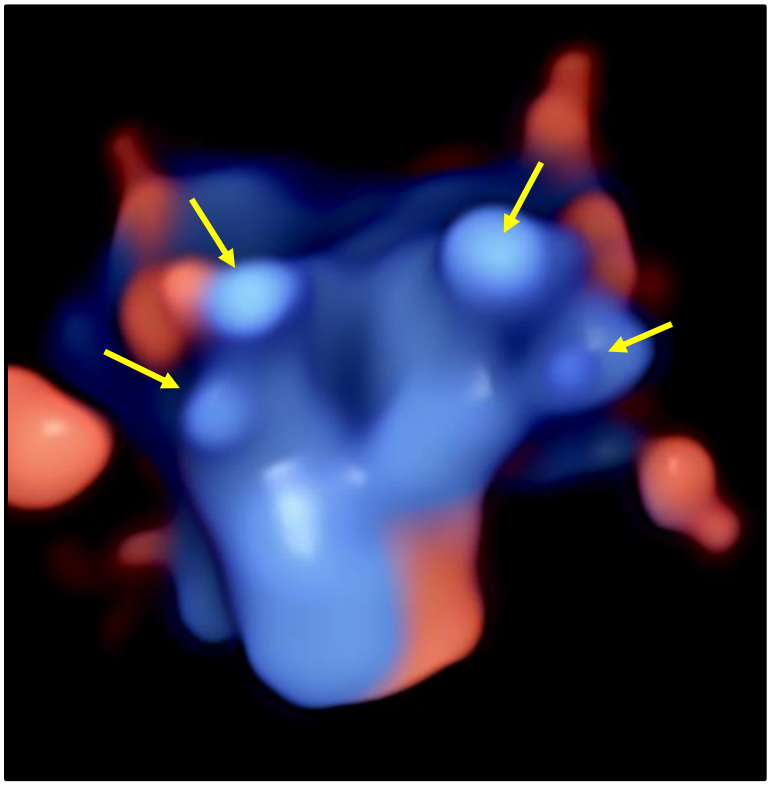


Fig.2: Double aortic arch. STIC color flow volume rendering showis each aortic arch giving off two vessels (arrows).

ABERRANT RIGHT SUBCLAVIAN ARTERY

Left aortic arch with an aberrant right subclavian artery (ARSA) is the most common aortic arch anomaly. ARSA arises from the distal part of the aortic arch and courses from the left side, behind the trachea and the esophagus, to the right upper arm. This is considered a normal variant, present in about 1% of the normal population.

ULTRASOUND FEATURES

- Three-vessel-trachea view on color Doppler shows ARSA arising from the isthmic region of the aortic arch, passing between the trachea and the spine, with a straight course towards the right arm (Fig.1).

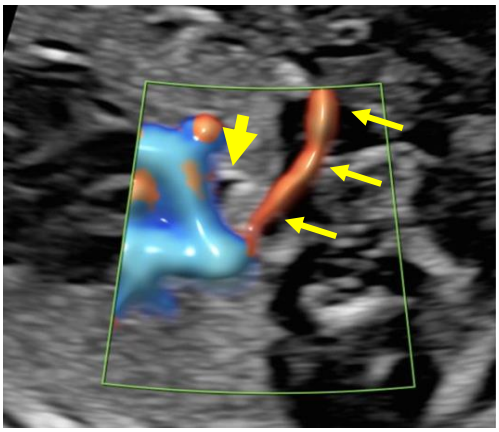


Fig. 1: Three-vessel-trachea view on color Doppler showing ARSA (small arrows) coursing behind the trachea (large arrow).

DIFFERENTIAL DIAGNOSIS

- Azygous vein- Drains into the superior vena cava, shows venous waveform on pulse wave Doppler.

ADDITIONAL POINTS

- Normal right subclavian artery in the axial plane is visualized as an S-shaped vessel passing anterior to the trachea (Fig.2 a).
- Color Doppler velocity settings should be adjusted downward (10 to 15 cm/sec) to facilitate the visualization of the small vessels.
- In cases with ARSA, the right carotid artery (RCA) or the right subclavian vein, which run anterior to the trachea, should not be mistaken for the normal right subclavian artery.
- ARSA is more common in fetuses with trisomy 21. Hence, it is considered an important soft marker for second-trimester aneuploidy screening. Additional soft markers or abnormalities are present in nearly all cases.
- ARSA with an absent or hypoplastic thymus may be markers for 22q11 deletion (Fig. 3).

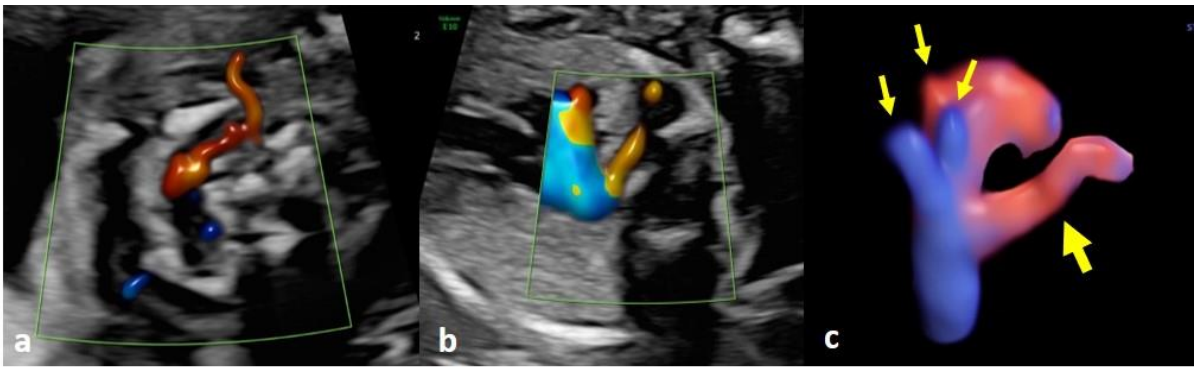


Fig. 2: (a) Normal S-shaped course of the right subclavian artery (bicycle handle appearance) (b) ARSA with origin from the isthmic region and a retrotracheal straight course (c) STIC volume in HD-flow with glass-body rendering shows three branches arising from the aortic arch (small arrows) and ARSA arising as the fourth branch (large arrow).

- In cases with ARSA as the only finding, there is insufficient evidence to recommend invasive testing. However, if the background risk is high or additional markers are seen, invasive testing should be advised to assess for trisomy 21 and 22q11 deletion.

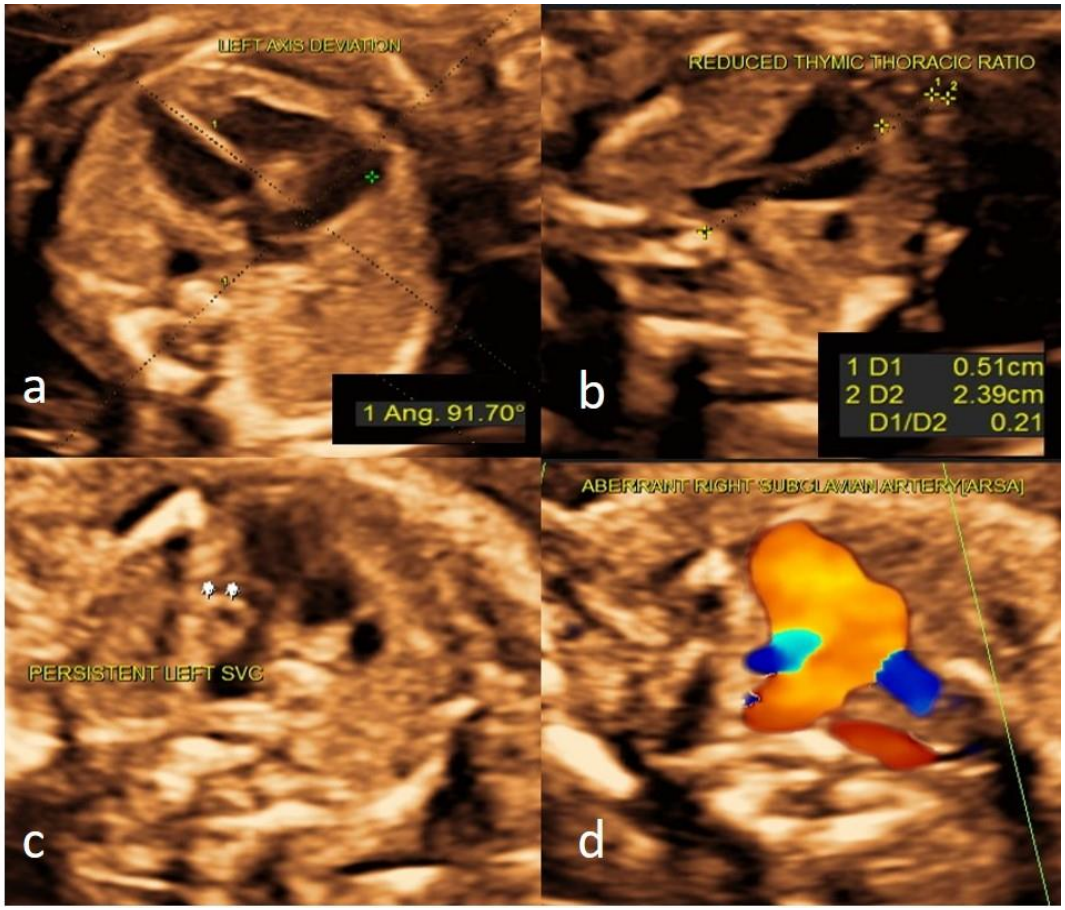


Fig. 3: 22 weeks 4 days fetus with (a) Left axis deviation of the fetal heart (b) Small sized thymus and reduced thymic-thoracic ratio (c) Persistent left superior vena cava (d) ARSA. No other cardiac or great vessel abnormality was found. Invasive testing with microarray was performed, which revealed 22q11 deletion. ***Case contributed by Dr Navya N, Bangalore***

ISOMERISM

RIGHT ATRIAL ISOMERISM

Right atrial isomerism (RAI) is a condition associated with the presence of “double” right-sided structures and the underdevelopment or absence of the left sided structures.

ULTRASOUND FEATURES

- Often presents with situs ambiguous, in which the fetal stomach and the cardiac apex are not on the same side.
- Upper abdomen shows a large midline liver with a right-sided or left-sided stomach.
- The position of the inferior vena cava (IVC) is anterior and on the same side of the descending aorta (juxtaposition of the aorta and IVC) (Fig.1a). This is considered a reliable sign for RAI.
- Stomach is usually displaced posteriorly in the abdomen (Fig.1b). This is due to asplenia. Stomach can also be positioned in the midline (Fig. 2 a).
- Both atria show broad based pyramidal shaped right atrial appendages.
- Nearly all cases show cardiac defects, usually complex cardiac anomalies. Many cases show unbalanced atrio-ventricular septal defect (Fig.1 c) with dominance of one chamber.
- Abnormal ventriculo-arterial connections, malposition of the great vessels with associated pulmonary stenosis or atresia are also common (Figs.1 d, e,and 2c).
- Right atrial isomerism is also commonly associated with partial or total anomalous pulmonary venous connections (Figs.1f and 2b), as the anatomic left atrium is absent.

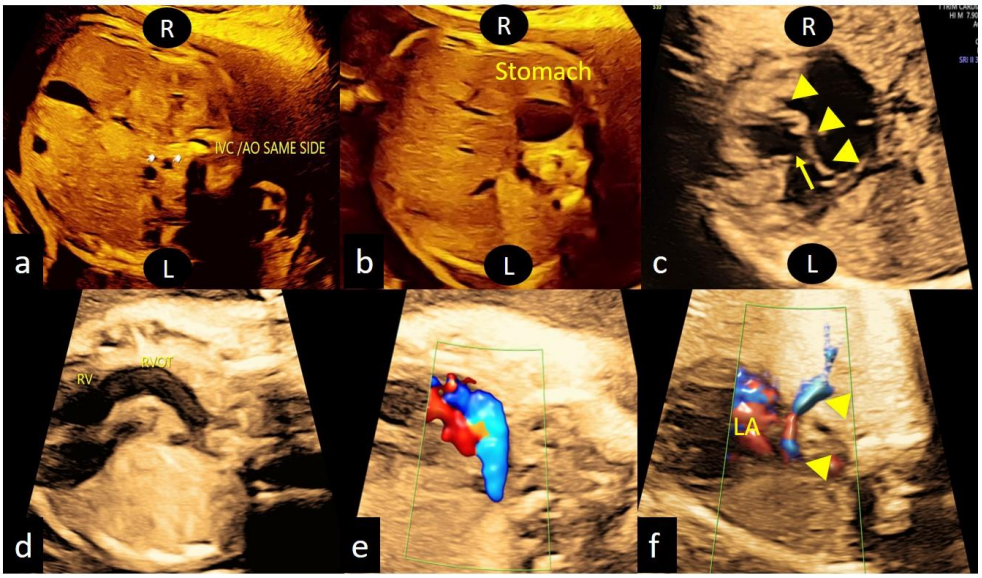


Fig. 1: Fetus in cephalic presentation (a) Juxtaposed aorta and IVC (b) Stomach on the right side and a large midline liver (c) Cardiac apex towards the left side. An atrioventricular septal defect (arrow) and linear insertion of the atrio-ventricular valves (arrowheads) (d) Double outlet right ventricle with a malposed aorta arising from the right ventricle (e) Reverse flow in the pulmonary artery (in red) due to pulmonary atresia (f) Anomalous pulmonary venous connection – the pulmonary veins are not draining into the left atrium.
Case contributed by Dr Bhagwan Shinde, Kopargaon

DIFFERENTIAL DIAGNOSIS

- Situs Inversus with dextrocardia
- Situs Inversus with levocardia

ADDITIONAL POINTS

- Associated extracardiac anomalies are frequent, mostly gastro-intestinal anomalies such as bowel atresia, bowel malrotation, diaphragmatic hernia, etc.
- Prognosis of right atrial isomerism is poor compared to left atrial isomerism due to presence of complex cardiac disease, extracardiac anomalies and neonatal sepsis due to asplenia.

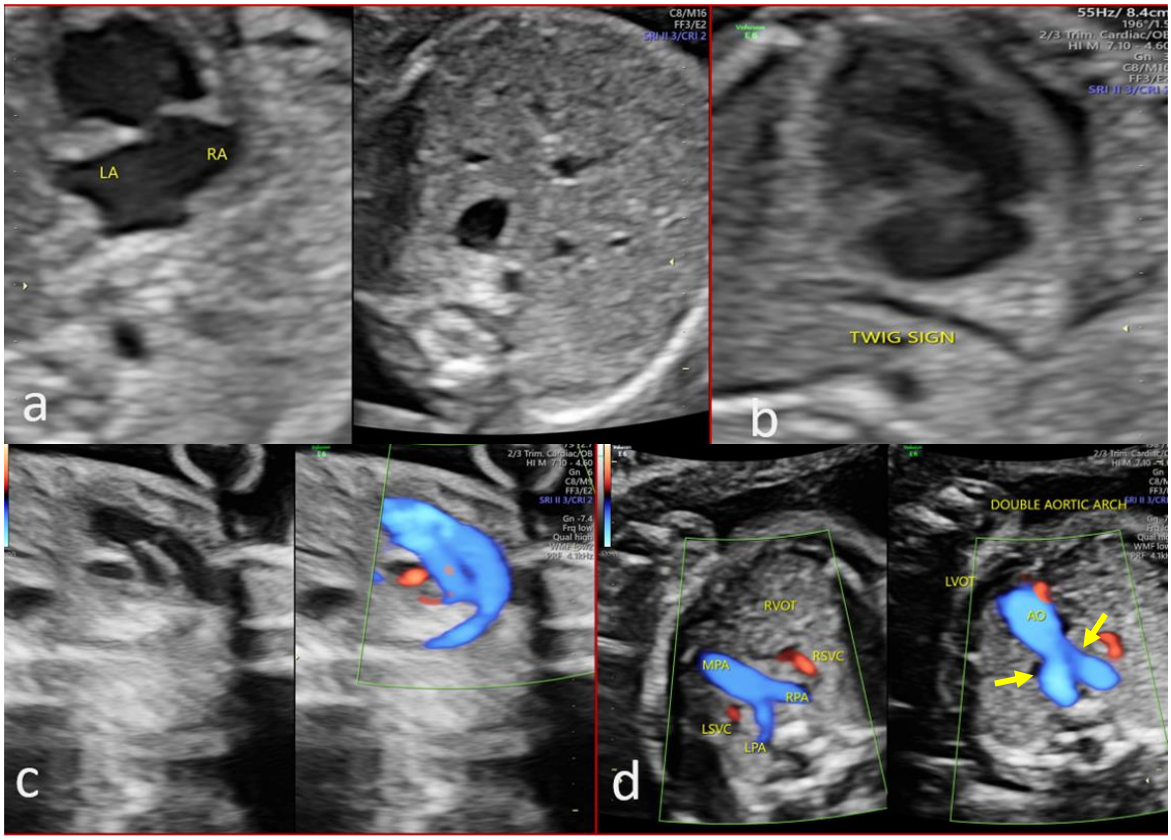


Fig2: (a) Four chamber view-A large monoventricle and situs ambiguous with a centrally placed stomach (b) Area behind the heart shows a confluent vein (twig sign).(c). LVOT view shows aorta and pulmonary artery arising from the right ventricle (Double outlet right ventricle) (d) Persistent left superior vena cava and double aortic arch (bifurcating ascending aorta, marked by arrows).

LEFT ATRIAL ISOMERISM

Left atrial isomerism (LAI) is a condition where there is the presence of double left-sided structures with underdeveloped or absent right-sided structures.

ULTRASOUND FEATURES

- Situs may be normal (Figs. 1 a and b). May present with a situs ambiguous or right-sided stomach (Figs.2a and b).
- The commonest finding in LAI is interrupted Inferior vena cava (IVC). The venous return from the lower part of the body is channelled via azygos or hemiazygos vein, increasing their caliber.
- Four-chamber view shows two vessels in the area behind the heart (Fig. 1b), which represent descending aorta and dilated azygos vein / hemiazygos vein.
- Parasagittal view of chest and abdomen shows azygos / hemizygous vein and descending aorta (double-vessel sign), which show opposite direction of flow (Fig. 1c).
- Complex cardiac anomalies, most notably atrioventricular septal defect (Fig.2a), may be seen. However, in many cases there are no complex malformations.
- Bradyarrythmia due to complete heart block is a frequent association (Fig. 2c).
- Both atria show hook-shaped atrial appendages.
- Outflow tracts are generally concordant and may show obstruction.
- Pulmonary venous anomalies are rare.
- Persistent left superior vena cava (PLSVC) is common (Fig. 1e).

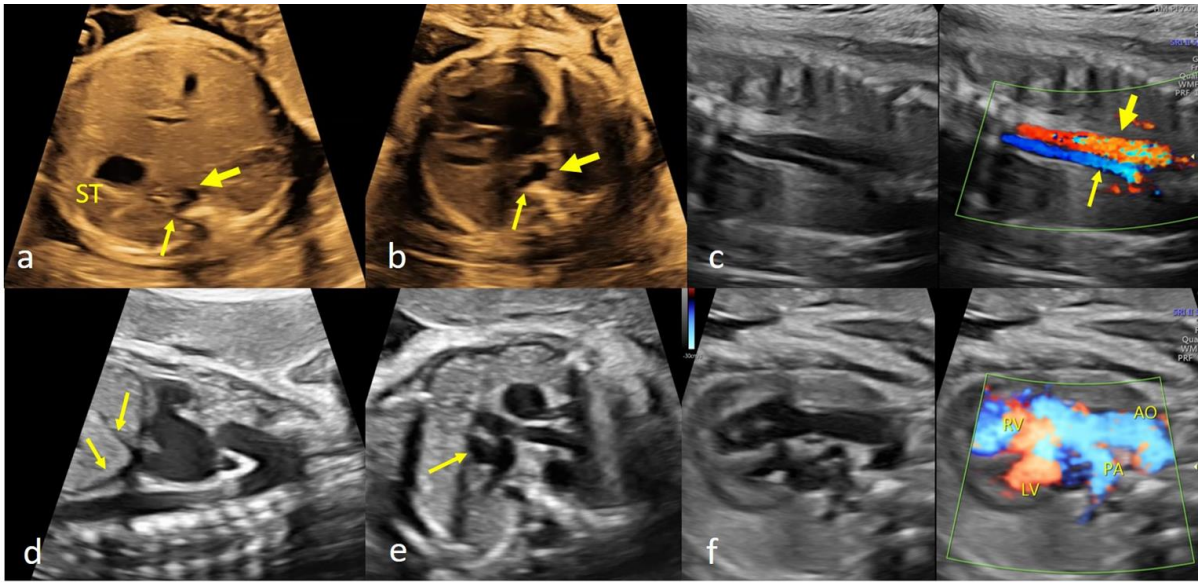


Fig.1: Left atrial isomerism. Fetus in cephalic presentation with situs solitus (a & b) Stomach (ST) and cardiac apex on the left side. Note two vessels close to each other in upper abdomen and chest, which are the aorta (thick arrow) and the hemiazygos vein on the left side of the aorta (thin arrow) (c) Parasagittal view of the abdomen shows the hemizygous vein (thin arrow) and the descending aorta (thick arrow), which show opposite direction of flow (d) The hepatic veins draining directly in the right atrium (e) Persistent left superior vena cava (arrow) (f) Complex cardiac malformation – Double outlet right ventricle

DIFFERENTIAL DIAGNOSIS

- Situs inversus with dextrocardia or levocardia
- Atrioventricular septal defect
- Dextrocardia with corrected transposition of great vessels

ADDITIONAL POINTS

- Associated extracardiac anomalies include mostly gastro-intestinal anomalies such as bowel atresia and malrotation, etc.
- LAI with no or a mild form of cardiac anomaly has a good prognosis.
- LAI with complete heart block and complex cardiac malformation has a poor prognosis. Intrauterine course may be complicated by hydrops.

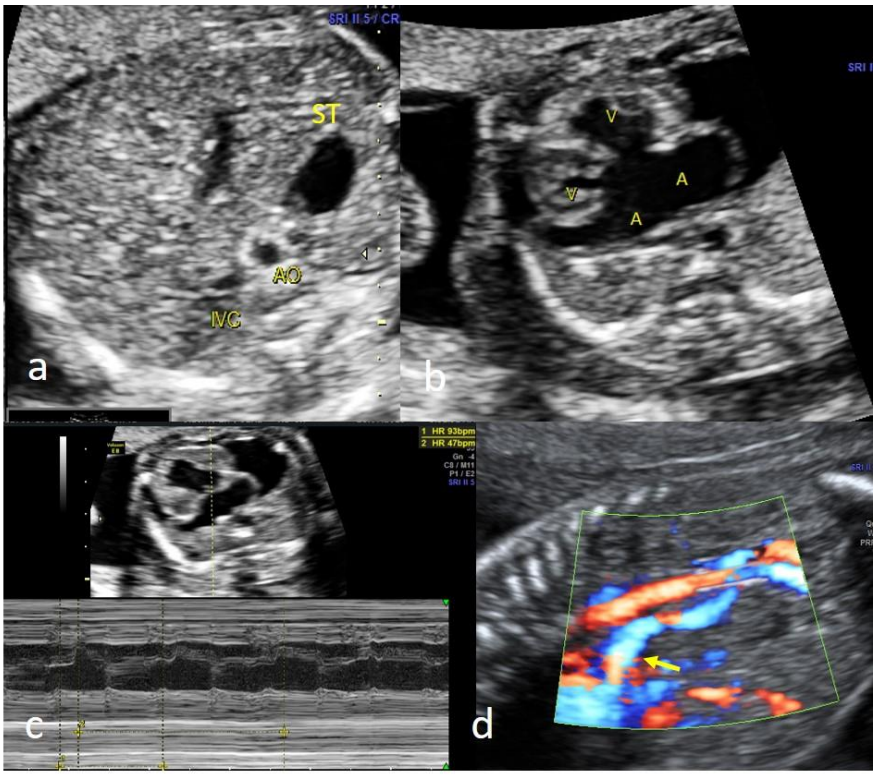


Fig. 2: Left atrial isomerism. Fetus in cephalic presentation with situs ambiguous (a) Stomach (ST) on the right side (b) Cardiac apex on the left side. Unbalanced atrioventricular septal defect (c) M-Mode shows bradycardia with a dissociation of the atrial and ventricular rates (complete heart block) (d) Parasagittal view of the abdomen shows the inferior vena cava draining in the right atrium (arrow)

UNIVENTRICULAR HEART

DOUBLE INLET SINGLE VENTRICLE

Double inlet single ventricle (DISV) is group of cardiac malformations in which two normally developed right and left atria connect via separate right and left atrioventricular valves to a common ventricle. The most common form of DISV, representing about 80% of cases, is a double inlet to a morphologic left ventricle (LV), also called **double inlet left ventricle (DILV)**. In DILV, a small underdeveloped right ventricle (RV) is commonly present and connects to the single ventricle via a ventricular septal defect, called as the **bulboventricular foramen**. The great arteries are often malposed.

ULTRASOUND FEATURES

- Single ventricle with a missing ventricular septum.
- In the single ventricle of the left ventricular morphology, the ventricular myocardium appears smooth with fine trabeculations (Fig. 1). When the morphology is right ventricle type, the ventricular myocardium is coarse with an irregular surface.
- A rudimentary ventricle, which is a small outlet chamber, is located on the left side of the main ventricle in L- Looping (Fig. 2) and on the right side of the main ventricle in D-Looping.
- The great arteries are generally in L-malposition if the small outlet chamber is on the left side of the main ventricle (Fig.2).

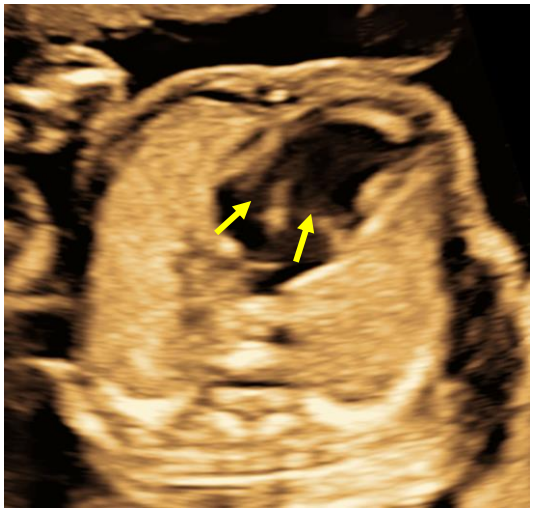


Fig.1: Two atrioventricular valves (arrows) opening in a single ventricle. The ventricle has smooth walls suggestive of left ventricle morphology

- When the small outlet chamber is located on the right side, the great arteries commonly arise in D-malposition. Rarely, the great arteries can be normally related; the pulmonary artery arises from the small outlet chamber **(Holmes heart).**
- The great artery which arises from the rudimentary outlet chamber is commonly obstructed. A narrow pulmonary artery suggests pulmonary stenosis or atresia, whereas a narrow ascending aorta suggests coarctation of the aorta or tubular aortic arch hypoplasia (Fig. 3).

DIFFERENTIAL DIAGNOSIS

- Hypoplastic left heart syndrome
- Mitral atresia with ventricular septal defect
- Pulmonary atresia with intact septum
- Tricuspid atresia
- Atrioventricular septal defect (unbalanced)

ADDITIONAL POINTS

- Diagnosis of DISV is typically made on gray-scale ultrasound.
- Color Doppler at the level of the four-chamber view may be misleading because two atrioventricular valves are patent and two color-stripes are visualized, thus mimicking the virtual presence of a separation or a septum.
- Color Doppler is often used for assessing patency of atrioventricular valves
- Commonly associated defects include atresia or hypoplasia of atrioventricular valves or semilunar valves and conduction abnormalities.
- Right or left atrial isomerism should be ruled out in a case with a single ventricle.

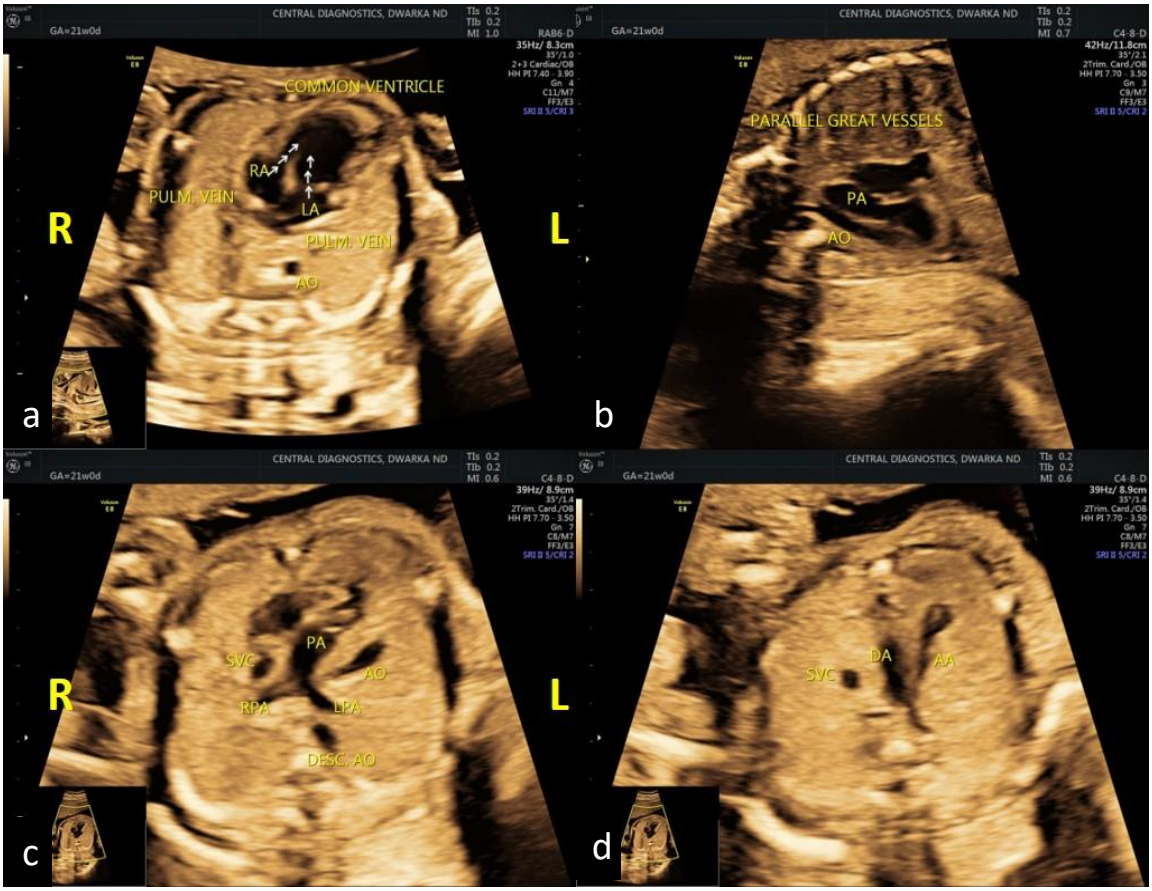


Fig.2: Double inlet single ventricle (a) Four-chamber view shows two atrioventricular valves (arrows) opening in a common ventricle (b) Sagittal view showing parallel great vessels. The aorta is arising from the rudimentary chamber, which is anterior and to the left of the single ventricle (L-looped ventricles) (c &d) Three-vessel and three-vessel-trachea views show malposed great vessels. Narrow caliber aorta is seen on the left side of a normal caliber pulmonary artery (L-malposition)

VENOUS ANOMALIES

TOTAL ANOMALOUS PULMONARY VENOUS CONNECTIONS

Total anomalous pulmonary venous connection (TAPVC) is a group of cardiac malformations in which all four pulmonary veins drain either directly into the right atrium or indirectly via a connection to the systemic venous system. TAPVC is a rare anomaly that often escapes prenatal detection. There are four types of TAPVC: type I- **supracardiac**, type II-**cardiac**, type III-**infracardiac** and type IV-**mixed pattern**. Supracardiac anomalous pulmonary connection is the most common and accounts for about 50% of cases, followed by infracardiac (25%), cardiac (16%), and mixed type (9%).

ULTRASOUND FEATURES

- Lack of a visible connection of the pulmonary veins to the left atrium (LA).
- Smaller LA and left ventricle (LV) on four-chamber view.
- Oval-shaped small LA with a smooth wall.
- Venous confluence behind the LA **(Twig sign)**.
- Abnormal, less pulsatile waveform in the pulmonary veins on Doppler.
- Increased area behind the heart (distance between the LA and the descending aorta). Distance more than the diameter of the descending aorta is an indirect sign for TAPVC.
- **Supracardiac type** (Fig. 1): vertical vein as a fourth vessel in the three-vessel-trachea view Dilated brachiocephalic vein and/or dilated superior vena cava (SVC).

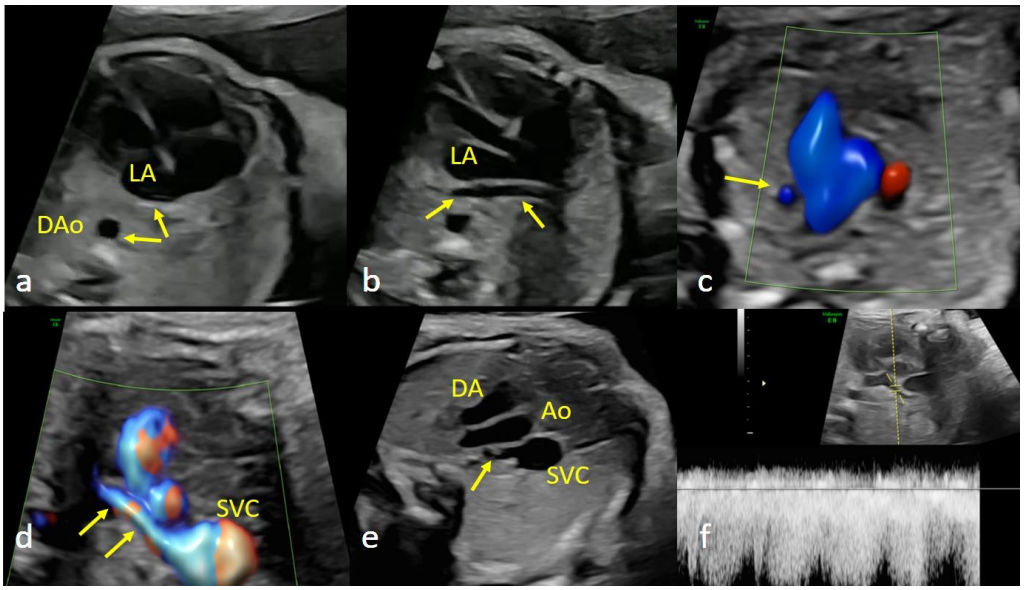


Fig.1: Supracardiac type TAPVC (a) Four-chamber view shows small LV and LA, smooth posterior wall of the LA and increased distance between the LA and the descending aorta (arrows)(b) Confluent vein behind the LA (c) Three-vessel-trachea view shows a dilated SVC (in red) and a vertical vein (in blue, marked by an arrow) to left of the pulmonary artery. The vertical vein has cardiofugal flow (away from the heart) and the SVC cardiopetal flow Hence, these veins show opposite flow direction (d & e) Vertical vein draining directly into the SVC (f) Spectral Doppler showing abnormal waveform in the pulmonary vein.

- **Cardiac type** (Fig. 2): The pulmonary veins drain into a dilated coronary sinus. Dilated coronary sinus in the absence of persistent left superior vena cava should caution us to look for a TAPVC.

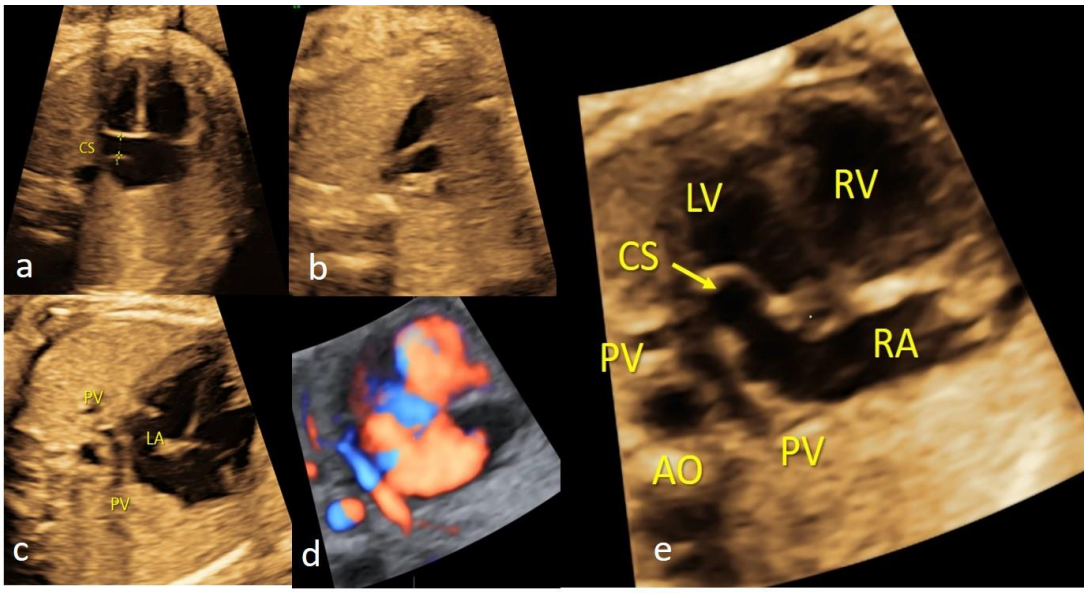


Fig.2: Cardiac type TAPVC (a) Four-chamber view shows a dilated coronary sinus (CS) (b) Three-vessel-trachea view shows no PLSVC (c & d) Pulmonary veins (PV) are seen behind the left atrium but not entering it (e) Pulmonary veins (PV) draining into the dilated coronary sinus (CS)

- **Infracardiac type** (Fig. 3): An additional vessel in the liver in cross-section. Vertical vein descending into the liver through the diaphragm from the chest.

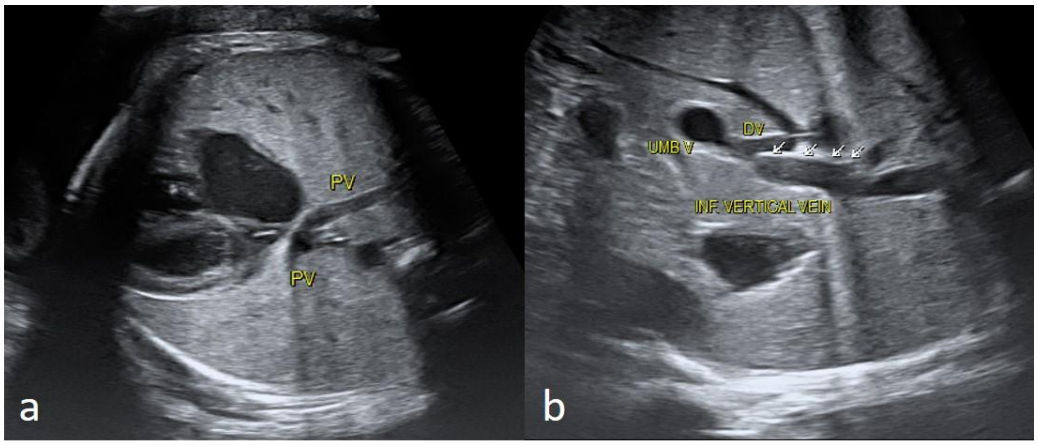


Fig.3: Infracardiac type TAPVC (a) Pulmonary veins (PV) not draining into the left atrium (b) Infracardiac vertical vein draining into the ductus venosus (DV)

DIFFERENTIAL DIAGNOSIS

- Interrupted inferior vena cava with azygos continuation: results in a dilated azygos vein posterior to the LA, which may be confused with a confluent vein.
- Persistent left superior vena cava (PLSVC) - imaged at the three-vessel-trachea view, may present similar to a supracardiac-type TAPVC. However, the flow direction in PLSVC is towards the heart, and the left brachiocephalic vein is absent.

ADDITIONAL POINTS

- Commonly associated cardiac defects include heterotaxy, primarily right atrial appendages isomerism, atrial septal defect (ASD) of sinus venosus type, (which is rarely detected prenatally), atrioventricular septal defect (AVSD), single ventricle, coarctation of Aorta (CoA), hypoplastic left heart syndrome (HLHS).
- A major prognostic factor in TAPVC is the presence of obstruction to the pulmonary venous return, which can cause a neonatal emergency warranting immediate surgery. Accurate prediction of obstruction of the pulmonary venous return can be difficult prenatally.
- Spectral doppler examination revealing peak velocities of greater than 50 cm/sec and/ or loss of pulsatile flow in the pulmonary veins or the vertical vein are the signs of a possibly obstructed TAPVC.

PERSISTENT LEFT SUPERIOR VENA CAVA

Persistent left superior vena cava (PLSVC) is the most common thoracic systemic venous anomaly, seen as a venous channel in the left neck and chest. PLSVC starts at the junction of the left jugular and subclavian veins, runs anterior to the ductal and aortic arches, and drains in the coronary sinus, which is located in a groove at the junction of the left atrium and ventricle.

ULTRASOUND FEATURES

- PLSVC is visualized in three-vessel or three-vessel-trachea view as a fourth vessel located left and anterior to the pulmonary artery and ductus arteriosus (Fig.1 a).
- A more cranial axial view of the mediastinum shows right and left superior vena cava along with a nearly always absent left brachiocephalic vein.
- Dilated coronary sinus is seen in an axial plane slightly caudal to the four-chamber view as a thin-walled bleb lateral to the mitral valve (Fig.1 b). A more caudal plane shows the coronary sinus as a channel in the left atrium, perpendicular to the interatrial septum.
- Left parasagittal view of the neck and chest can also demonstrate the PLSVC as a linear channel draining into the coronary sinus (Fig.1 c).
- Rarely, the right superior vena cava (RSVC) may be absent. This results in visualization of three vessels in mediastinal planes (Fig. 2). The brachiocephalic vein is present in such cases and the PLSVC appears more dilated.
- Color Doppler shows flow direction towards the heart in the PLSVC. The coronary sinus shows flow direction from left to right, opposite to the flow across the foramen ovale.

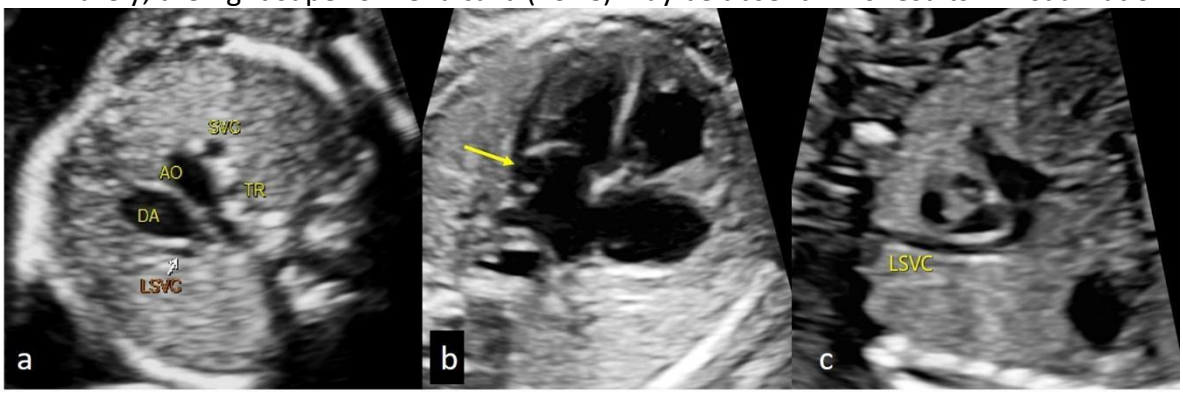


Fig. 1: PLSVC (a) Four vessels in the three-vessel-trachea view due to persistent left superior vena cava (LSVC) (b) Dilated coronary sinus (arrow) in the left atrial wall (c) Parasagittal view of the chest showing PLSVC as a linear channel draining into the coronary sinus

DIFFERENTIAL DIAGNOSIS

- Vertical vein of supracardiac total anomalous pulmonary venous connection (TAPVC): The direction of flow in the vertical vein is towards the head. The brachiocephalic vein is present, is of normal size or dilated.
- Redundant ductus arteriosus: usually seen in the third trimester, shows arterial flows and continuity with the ductus arteriosus.

ADDITIONAL POINTS

- PLSVC is thought to be the result of an in-utero failure of regression of the left anterior & common cardinal veins.
- In the majority of cases, PLSVC has no clinically significant hemodynamic implications.
- Rarely, PLSVC may drain into the left atrium, either directly or via an unroofed coronary sinus. This causes a right-to-left cardiac shunt.
- A common association of PLSVC is aortic coarctation. Hence, in cases with PLSVC, the left ventricular and aortic arch diameters should be monitored.
- Other heart abnormalities such as right aortic arch (Fig. 3), cono-truncal abnormalities, and heterotaxy syndromes may also have PLSVC as an associated finding
- Rarely, chromosomal abnormalities such as trisomy 21 and 18 may be found, especially when other cardiac or extracardiac anomalies are present. However, in isolated PLSVC, there is insufficient evidence to offer invasive prenatal testing.

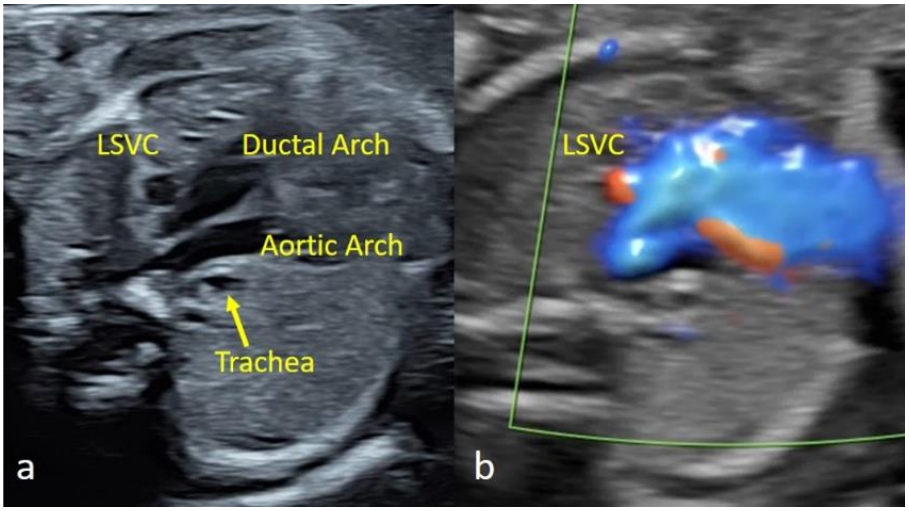


Fig. 2: PLSVC with absent right superior vena cava

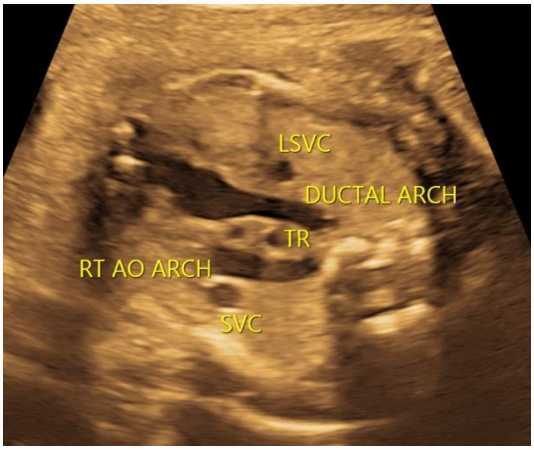


Fig. 3: PLSVC in a case of right aortic arch

MISCELLENEOUS

RHABDOMYOMA

Fetal cardiac tumors are rare; about 80–90% of these are Rhabdomyomas, which are benign mesenchymal tumors of striated muscle origin. These are generally detected between 20 to 30 weeks of gestation.

ULTRASOUND FEATURES

- Single or multiple round-to-oval shaped well-margined nodular lesions arising mostly from the ventricular myocardium (Fig. 1).
- Echogenic lesions with homogeneous echopattern with no calcification or cystic areas.
- Frequently protrude into the cardiac lumen and, rarely, may cause flow obstruction.

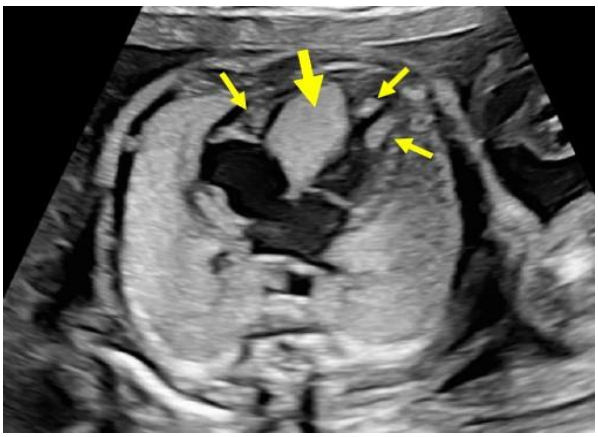


Fig. 1: Multiple Rhabdomyomas. A large echogenic mass in the right ventricle along the ventricular septum (large arrow). Other smaller lesions along the walls of both ventricles (small arrows).

DIFFERENTIAL DIAGNOSIS

- Fibroma: single, large mass and presents with pericardial effusion.
- Teratoma: heterogeneous large extracardiac mass with cystic areas and calcification.
- Myxoma: echogenic mobile lesion with a stalk.

ADDITIONAL POINTS

- Rhabdomyoma is usually the earliest sign of Tuberous Sclerosis in-utero.
- Tuberous Sclerosis is caused due to mutations of tumor suppressor genes- TSC1 and TSC2. These can be identified prenatally on genetic testing.
- The risk for Tuberous sclerosis is > 90% in cases with multiple Rhabdomyomas.
- A fetus with a seemingly solitary Rhabdomyomas may develop other smaller tumors as gestation progresses.
- Additional signs of Tuberous sclerosis in fetuses are cortical tubers, subependymal nodules, and renal angiomyolipoma.
- Cortical tubers and subependymal nodules in the fetal brain are best seen with TVS (Fig. 2 d).
- Small Rhabdomyomas do not impair cardiac activity and can regress after 30 weeks or postnatally.
- Larger cardiac tumors (>20 mm) are rare, can lead to hemodynamic impairment and arrythmia.

- Fetal MRI is an additional imaging modality to detect cranial and renal associations.

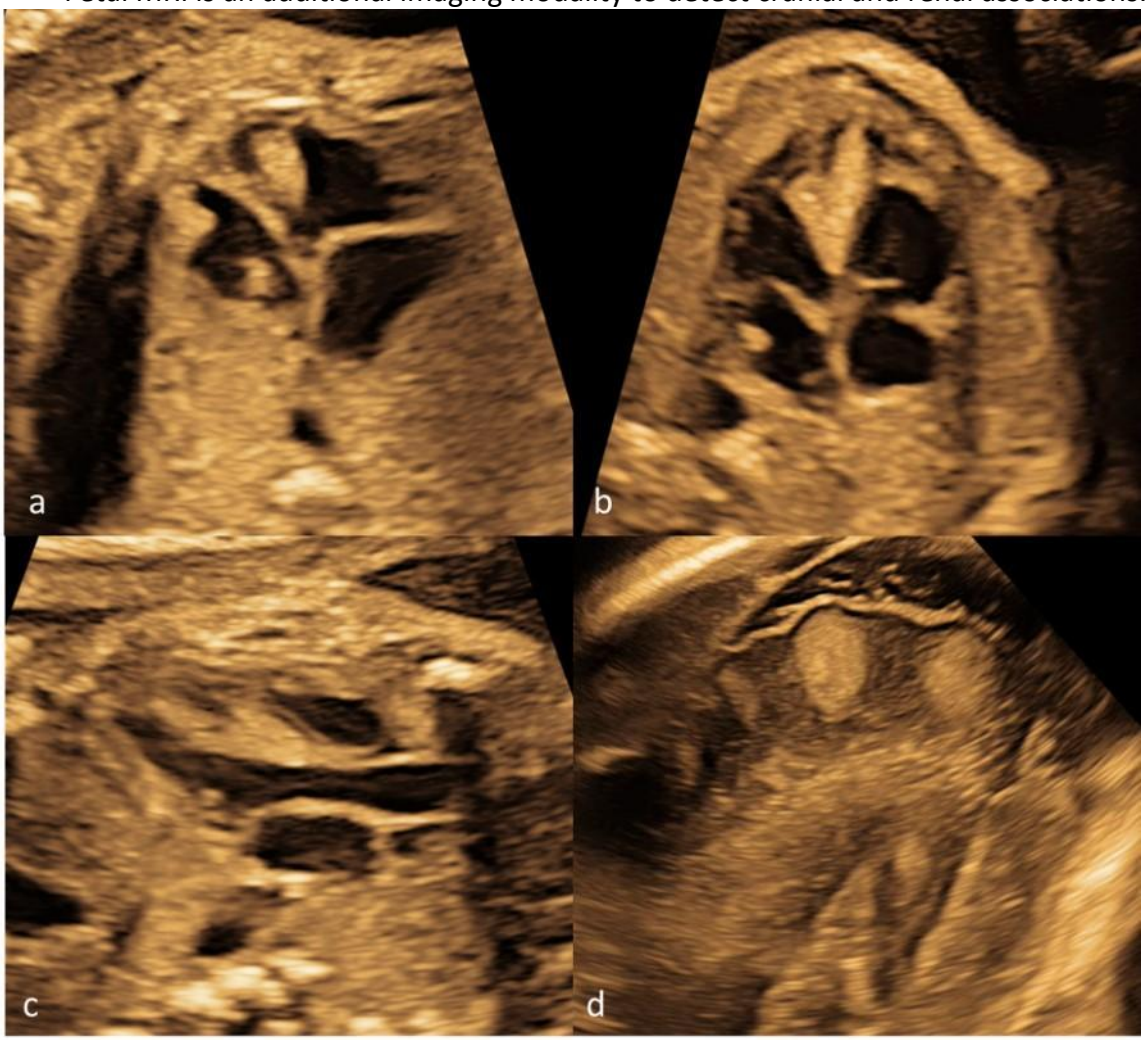


Fig 2: A case of Tuberous Sclerosis (a & b) Four chamber view- Multiple echogenic masses arising from both ventricular walls, left atrial wall and interventricular septum. (c) LVOT view- thickened and echogenic interventricular septum. (d) Parasagittal view of the brain- Multiple echogenic masses in cortex with BPD and HC > 95th centile suggestive of tubers.

Case contributed by Dr Sri Sai Lakshmi B J, Hyderabad

FETAL CARDIOMYOPATHIES

Cardiomyopathies are a heterogeneous group of diseases that affect the myocardium, usually without any underlying structural cardiac anomalies. These are associated with abnormal cardiac function, which can cause hydrops and fetal demise. Four types of cardiomyopathies are seen in fetuses : dilated, hypertrophic, restrictive, and ventricular non-compaction.

ULTRASOUND FEATURES

- **Dilated cardiomyopathy:** 1. Enlarged fetal heart (Fig. 1), either assessed subjectively or by cardiac measurements such as cardiac width, cardio-thoracic ratio or z-scores of the cardiac chamber size. 2. Poor contractility, assessed as poor fractional shortening on M-mode or abnormal myocardial performance index. 3. Atrio-ventricular valvular regurgitation. 4. Pericardial effusion 5. Hydrops.

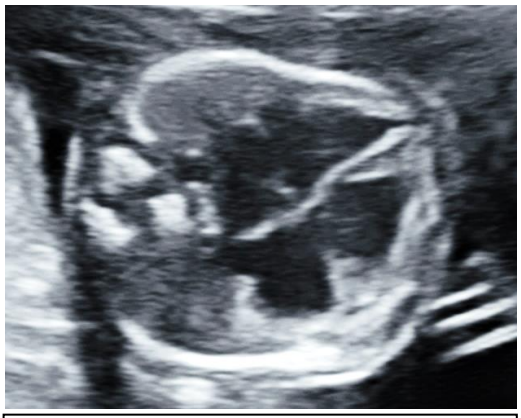


Fig 1: Cardiomegaly in a 19-week fetus The heart showed poor contractility. No obvious cause was found.

- **Hypertrophic cardiomyopathy:** 1. Enlarged heart associated with thickened ventricular walls (Fig. 2). Diastolic thickening > 2 z-scores is abnormal. 2. Reduced lumen of the ventricles 3. Thickened interventricular septum 4. Outflow tract obstruction may be found in some cases, seen as aliasing on color Doppler. 5. Pericardial effusion.
- **Restrictive cardiomyopathy:** Very rare, suspected when the ventricular size and wall thickness are normal but the atria are enlarged.
- **Ventricular non-compaction:** 1. Thickening of the ventricular walls. 2. Prominent trabeculations and deep recesses in the ventricular walls, giving them a spongy appearance (Fig. 4)

DIFFERENTIAL DIAGNOSIS

- Volume overload due to shunts, twin-to-twin transfusion and other causes: cardiac contractility and function are normal. Can progress to dilated cardiomyopathy.
- Hypoplastic left or right heart syndrome: thickened left or right ventricle, reduced ventricular cavity size and contractility. Associated with valvular atresia/ severe stenosis.
- Left ventricular endocardial fibroelastosis: thickening of the ventricular wall with increased echogenicity of the endocardium due to high grade stenosis of the aortic valve.
- Ebstein anomaly and tricuspid dysplasia: cardiomegaly, severe tricuspid regurgitation.

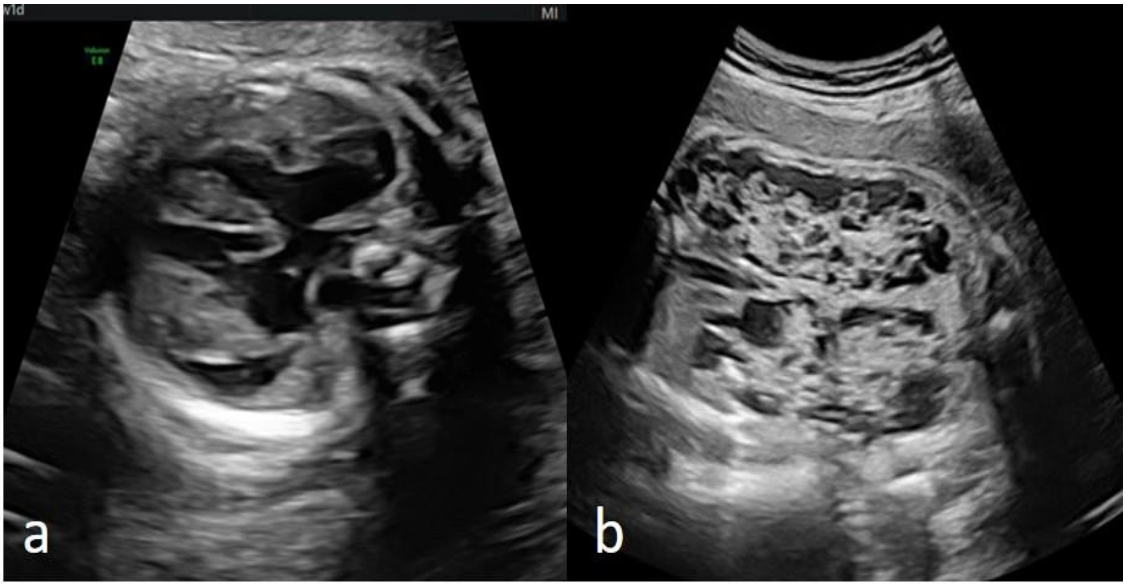


Fig 2: Hypertrophic cardiomyopathy in a case of Autosomal recessive polycystic kidney disease (a) Grossly dilated fetal heart filling the entire chest. The ventricles show diffusely thickened walls (b) Bilateral enlarged fetal kidneys with numerus cysts

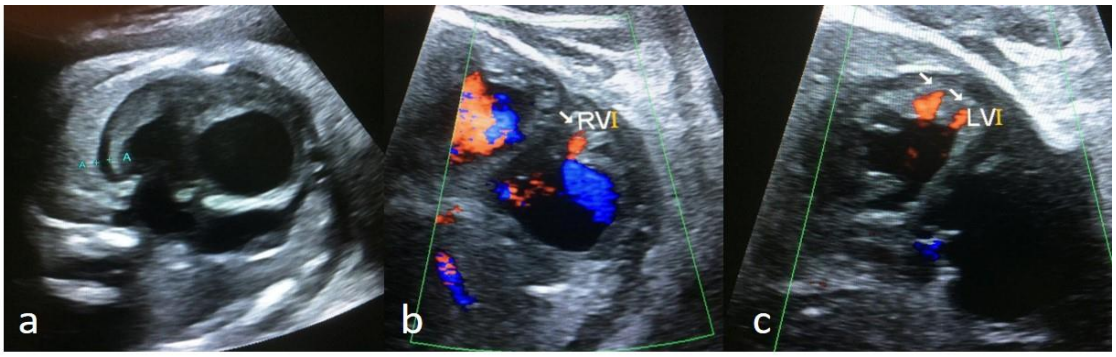


Fig 3: Ventricular non-compaction (a) Dilated fetal heart with thickened ventricular walls, most pronounced at the cardiac apex (b &c) Deep recesses in the myometrium of the right and left ventricular apices, showing color flow.
Case contributed by Dr Meenal Jain, Jodhpur

ADDITIONAL POINTS

- Cardiomyopathy may have an underlying infective, genetic, metabolic or feto-maternal cause.
- Dilated cardiomyopathy - A survey of the fetus and the placenta should be performed to look for any atrio-venous malformation or shunt. Signs of fetal anemia and irregular cardiac rhythm can suggest parvovirus B19 infection (Fig.4). Bradycardia and heart block may suggest maternal autoantibodies.
- Hypertrophic cardiomyopathy: Maternal diabetes, fetal bilateral renal agenesis or dysplasia and twin-twin transfusion are the common causes. Heart block may suggest maternal autoantibodies. Noonan syndrome and other rasopathies may be associated with hypertrophic cardiomyopathy.

- MYH7 gene is implicated in many forms of cardiomyopathies.
- Prognosis is variable. Many cases show progressive deterioration in fetal and postnatal life. Development of hydrops is a poor prognostic sign. Remarkably, some cases may show improvement, even complete resolution, particularly the cardiomyopathies associated with maternal diabetes and maternal autoimmune disorders post-treatment.

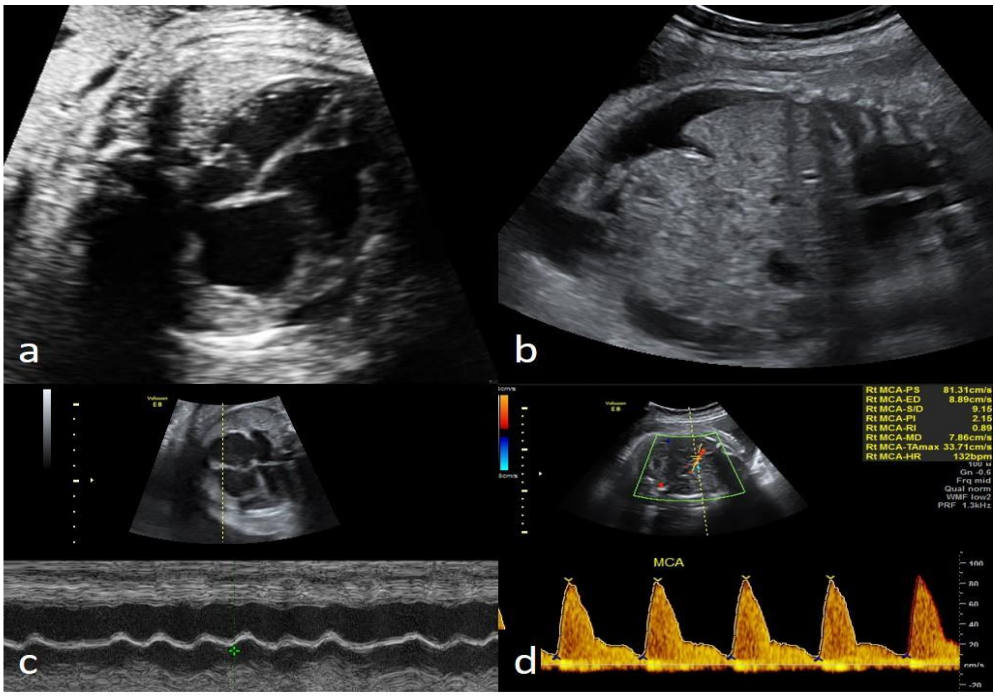


Fig.4: Dilated cardiomyopathy caused by Parvovirus B19 infection (a) Cardiomegaly (b) Fetal ascites and body wall edema as signs of hydrops (c) M-mode trace showing poor contractility of the heart and an irregular cardiac rhythm (d) Increased peak systolic flow velocity in fetal MCA >1.5 MoM suggestive of fetal anemia.

CORONARY CAMERAL FISTULA

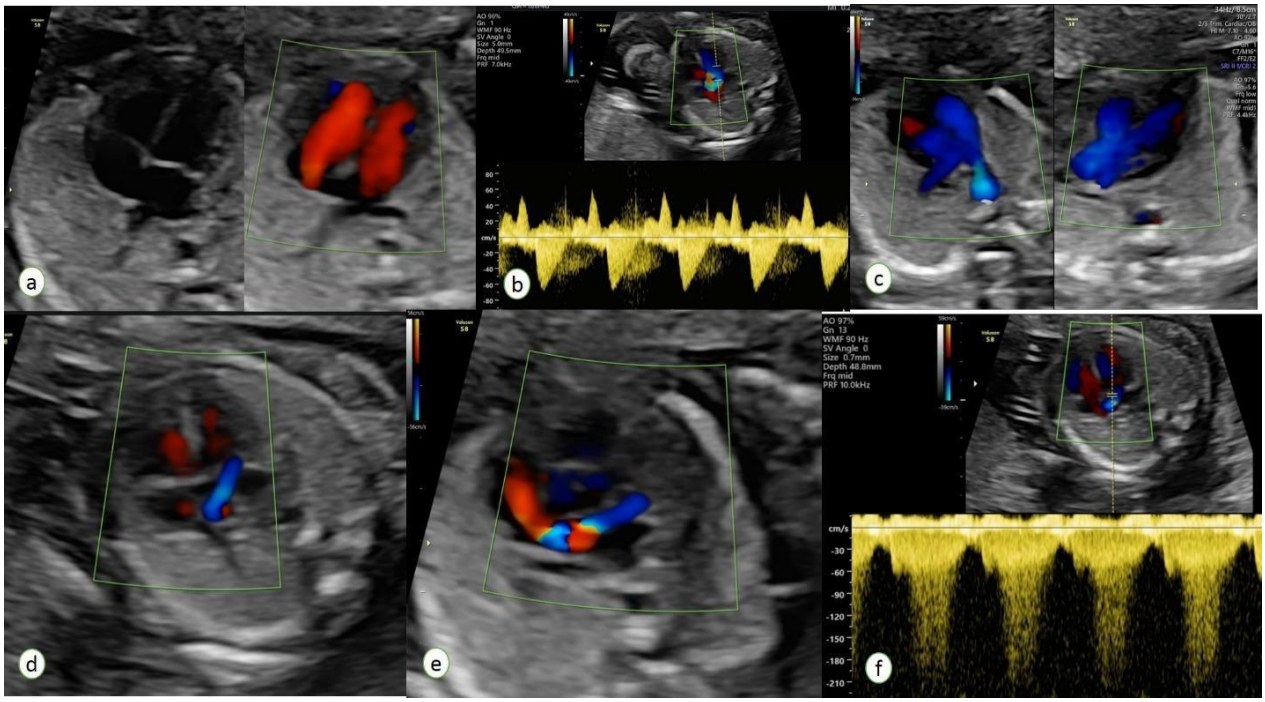


Fig. 1: (a) Normal four-chamber view of the heart; (b) Spectral Doppler across the mitral valve shows normal mitral flows with an additional mixing of a waveform from the coronary fistula; (c) RVOT and LVOT show normal antegrade flow. (d) High flow Coronary artery fistula extending from the base of the mitral valve and reaching up to the base of the right atrium (e) Jet of flow from the fistula into the right atrium; (f) Spectral Doppler showing high velocity flow in the coronary fistula. **Case contributed by Dr. Nitin Jadhav**

ULTRASOUND FEATURES

- Detection of isolated coronary artery fistula antenatally is very rare.
- Four-chamber view is normal.
- Outflow tracts are also of normal caliber and show antegrade flow.
- In this case the coronary artery fistula is seen extending from the base of the mitral valve (probably from the left coronary artery) into the right atrium.
- These fistulas usually show aliasing on color Doppler with high velocity flow on spectral Doppler.
- Different coronary artery fistulas are reported in postnatal series with their communication with different chambers of the heart as well as the outflow tracts.
- These fistulas may be associated with other abnormalities. In this case everything else was normal. The fistula was also demonstrated in the 32-week scan and confirmed on the postnatal echo.

FIRST TRIMESTER ANOMALIES

ATRIOVENTRICULAR SEPTAL DEFECT (AVSD)

AVSD is an abnormality characterized by a deficient AV (atrio-ventricular) septum and abnormal AV valves, resulting in a common AV junction.

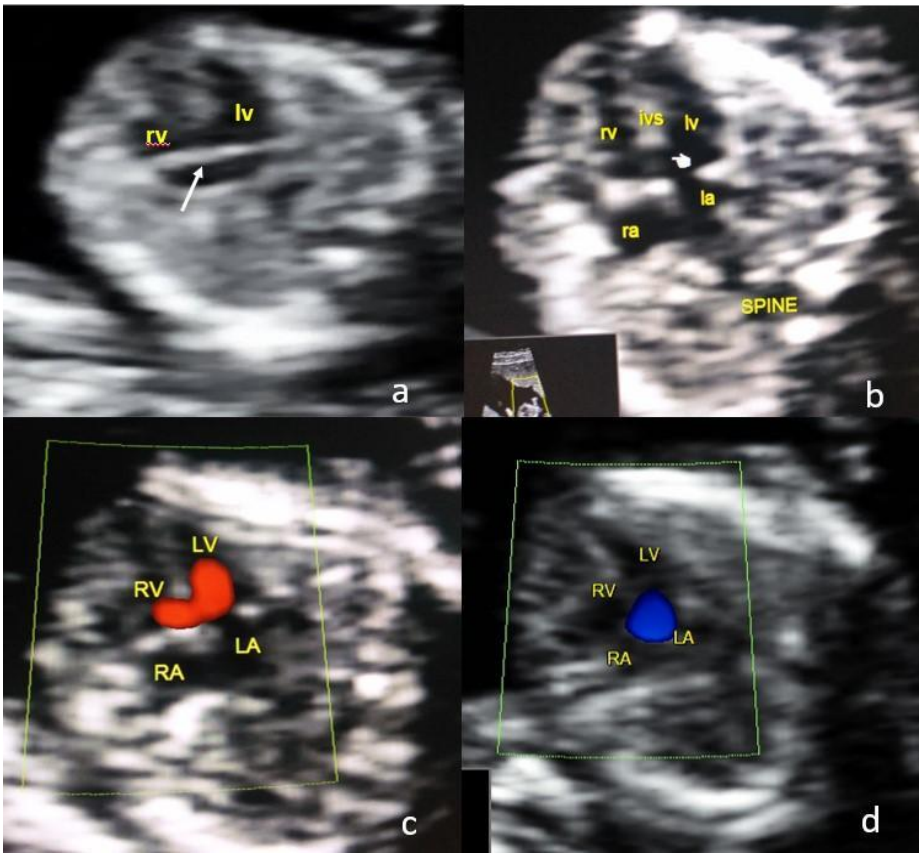


Fig.1: (a) Four-chamber view shows a linear insertion of the AV valves (arrow) in systole (b) central cardiac defect in diastole (c) colour Doppler shows flow across the septal defect (d) regurgitation at the common AV valve. The patient underwent invasive testing, which revealed trisomy 21.

ULTRASOUND FEATURES

- Four-chamber view shows linear insertion of the AV valves in systole (this may be difficult to detect in the first trimester), a central defect in the heart in diastole (Figs.1 a & b).
- Color Doppler shows a central column of blood in the common AV canal (Fig. d).
- Pulse-wave Doppler shows regurgitation at the AV valve.

DIFFERENTIAL DIAGNOSIS

- It is difficult to differentiate AVSD from the other entities in the first trimester. Based on color Doppler findings, a univentricular heart is a close differential. Follow up scan at 16weeks is indicated.

ADDITIONAL FINDINGS

- AVSD is associated with trisomy 21, and to a lesser extent with trisomy 18 and 13.
- Antenatal diagnosis of AVSD, when isolated, is associated with trisomy 21 in 58% of the cases.
- If AVSD is associated with heterotaxy, then the risk of a chromosomal abnormality is reduced. Prognosis is poor due to associated cardiac malformations.

HYPOPLASTIC LEFT HEART SYNDROME (HLHS)

HLHS is a complex anomaly with an underdeveloped left heart, resulting in a reduction of the systemic cardiac output. It manifests in two forms, 1. Atresia of aortic and mitral valves with connection between atria and ventricle 2. Aortic atresia with mitral dysplasia, resulting in a small left ventricle and a severely dysplastic mitral valve with aortic stenosis or atresia.

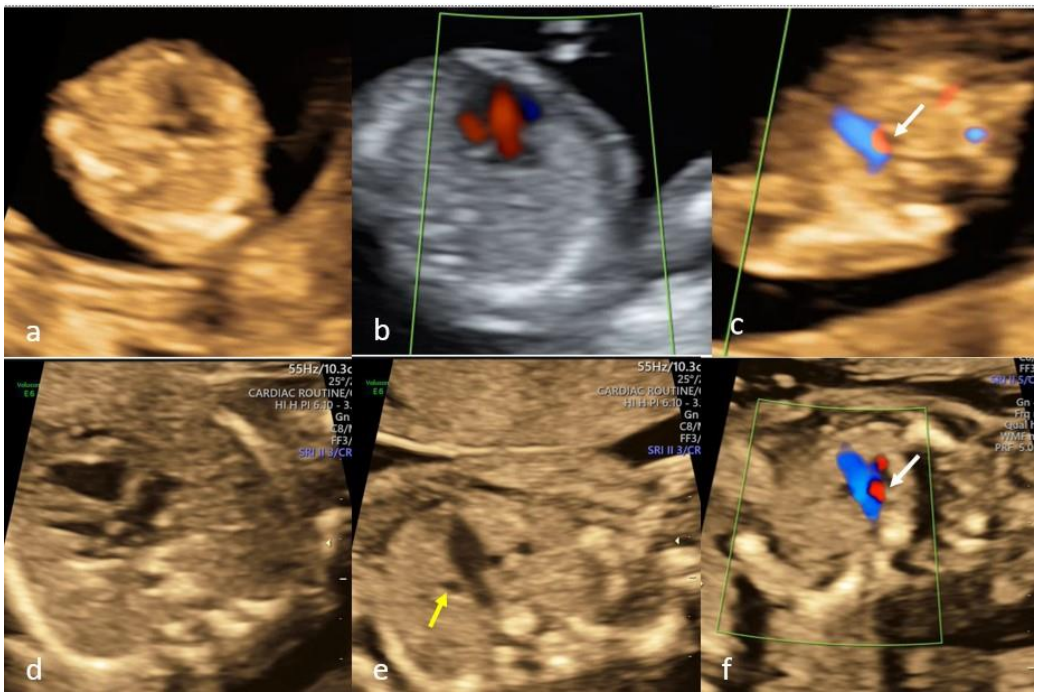


Fig.1: (a and b) Four-chamber view show a small left ventricle with reduced LV inflow. (c) Three-vessel-view shows forward flow in the pulmonary artery and reversed flow in the aortic arch (arrow). (d) Follow up scan at 16wks. Four-chamber view shows small left ventricle (e) Three-vessel-trachea view shows atretic aorta and a persistent left superior vena cava (arrow) (f) Three-vessel-trachea view shows flow reversal in the aorta (arrow).

ULTRASOUND FINDINGS

- Four-chamber view appears abnormal with a small left ventricle. In aortic and mitral atresia the left ventricle is absent (Figs. a and d).
- Color Doppler shows asymmetric ventricular inflow or a single channel ventricular inflow, with no flow in the left heart (Fig. b).
- Three-vessel-trachea view shows a small atretic aortic arch (Fig. e). Color Doppler shows a dilated pulmonary artery with forward flow and a small aortic arch with flow reversal (Figs.c and f).
- Echogenic left ventricle is seen in some cases in the first trimester, which represents endocardial fibroelastosis.

DIFFERENTIAL DIAGNOSIS

- There are many conditions showing asymmetric or single ventricle flow in the first trimester. Differentiating HLHS from other entities may be difficult in the first trimester. However, **Coarctation of aorta** is a close differential. The aortic arch in coarctation shows forward flow, unlike in HLHS, where the flow is reversed.

ADDITIONAL POINTS

- When HLHS is suspected, counseling regarding genetic testing should be performed, as it is associated with trisomies, Turner's and other syndromes.
- Fetuses with HLHS develop fetal growth restriction in the late second and third trimesters.
- The diagnosis of HLHS in the first trimester should be confirmed by follow up scans.

TRICUSPID ATRESIA

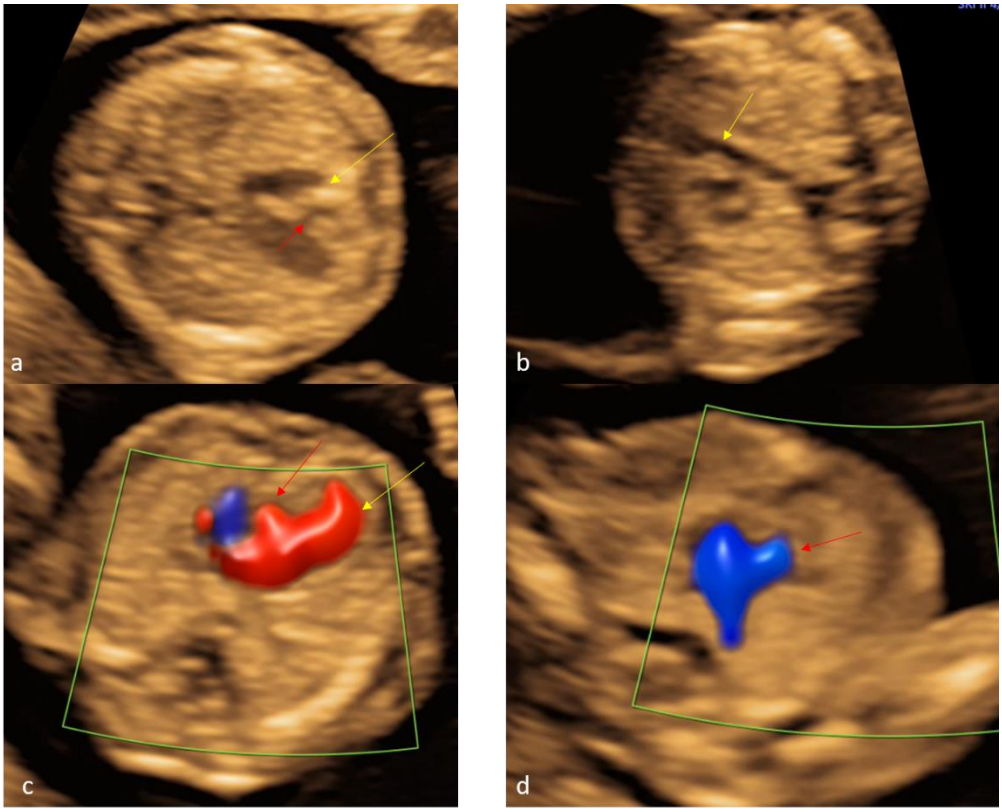


Fig 1: (a) Four-chamber view shows an echogenic immobile tricuspid valve (yellow arrow), a small right ventricle and a ventricular septal defect (red arrow). (b). Three-vessel view shows a small pulmonary artery (yellow arrow). (c). Four-chamber view with color Doppler shows normal filling of the left ventricle (yellow arrow). The right ventricle is filling through the VSD (red arrow). (d) Three-vessel-view with color Doppler shows a small pulmonary artery with forward flow.
Case *contributed by Dr Sri Sai Lakshmi B J, Hyderabad*

ULTRASOUND FEATURES

- Four-chamber view shows an echogenic immobile tricuspid valve and a small right ventricle. VSD is a common association.
- Three-vessel and three-vessel-trachea view show a small pulmonary artery.
- Color Doppler shows blood flow across the mitral valve with filling of the small RV through VSD. The pulmonary artery is small in caliber and shows forward flow.

DIFFERENTIAL DIAGNOSIS

- Pulmonary atresia with intact inter-ventricular septum
- Tricuspid stenosis
- Tetralogy of Fallot

ADDITIONAL POINTS

- Tricuspid atresia is a cyanotic congenital heart disease with complete agenesis of tricuspid valve.
- They are classified as Type I- normal great arterial connection; Type II- transposition of great arteries; Type III- with persistent Truncus arteriosus.
- Type I and II are subdivided into Subgroup a- with pulmonary atresia; Subgroup b – with pulmonary stenosis; Subgroup c- with absence of obstruction
- The amount of pulmonary blood flow depends on the degree of pulmonary obstruction, presence of VSD and the relationship of the great arteries.
- Tricuspid atresia is usually not associated with extracardiac defects or chromosomal aberrations.

PULMONARY ATRESIA WITH INTACT VENTRICULAR SEPTUM (PA-IVS)

PA-IVS is a condition with no communication between the right ventricle and the pulmonary artery with an intact interventricular septum.

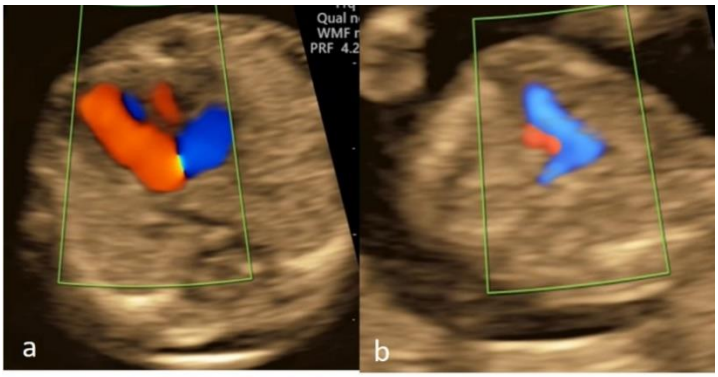


Fig.1: (a) shows asymmetric ventricular inflow with reduced flow in the right ventricle (b) Three-vessel view shows forward flow in the aorta and reversal in the ductus arteriosus.

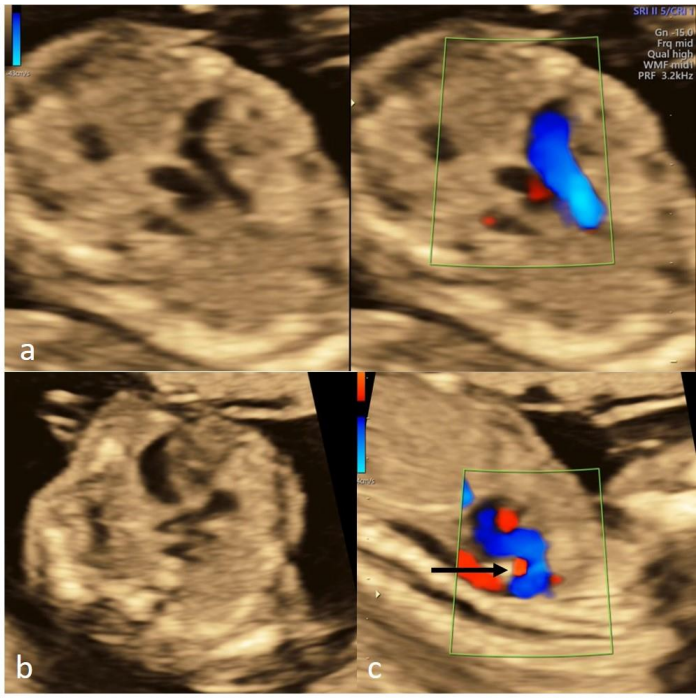


Fig 2: (a) shows normal left outflow tract with maintained septo-aortic angle (b). Four-chamber view shows the interventricular septum bulging in the left ventricle cavity (c) aortic arch view shows reversal of flow in the ductus arteriosus (black arrow)

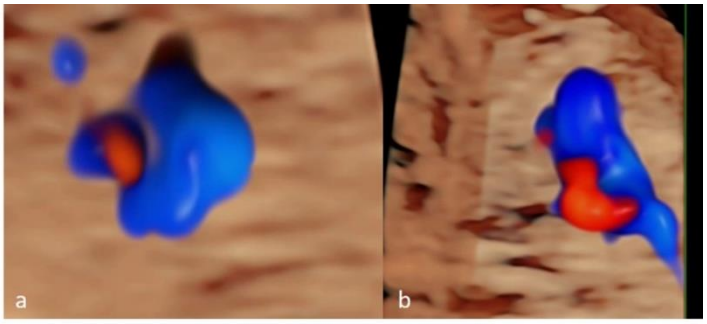


Fig. 3:4D STIC color flow volume rendering at 13wks (a) and at 16wks(b) shows forward flow in the aorta (blue) and flow reversal in the ductus arteriosus (red)

ULTRASOUND FEATURES

- Detection of this condition in early gestation is first suspected by asymmetric ventricular inflow, with reduced flow across the tricuspid valve (Fig.1a).
- Outflow tracts show forward flow in the aorta and reversal in the pulmonary artery (Fig. 1b).
- Aortic arch view shows reversal of flow in the ductus under the arch (Fig.2c).
- Other features like bulging interventricular septum (Fig.2b) and ventriculo-coronary connections (VCAC) are usually appreciated in the early to mid-trimester scans. In some cases these findings may be seen in early gestation.

DIFFERENTIAL DIAGNOSIS

- Tricuspid atresia with ventricular septal defect: color Doppler demonstrates no flow across the tricuspid valve, in contrast to PA-IVS, in which flow across the tricuspid valve is often seen.
- Pulmonary atresia with ventricular septal defect: in this condition there is a perimembranous ventricular septal defect (VSD) with an overriding aorta. The ventricles are of equal size in this entity. In later stages, major aorto-pulmonary collaterals (MAPCAS) can be demonstrated.
- Pulmonary stenosis: antegrade flow across the pulmonary valve is demonstrated on color Doppler.

ADDITIONAL POINTS

- Pulmonary stenosis with presence of a normal four-chamber view and normal diastolic filling can progress to pulmonary atresia.
- Ventriculo-coronary artery connections (VCACs) are found in about one-third of the cases of PA-IVS.
- In four-chamber view, the right ventricle can be hypoplastic or normal.
- Associated chromosomal abnormality with PA-IVS is rare.
- Prognosis is dependent on the size and function of the right ventricle.

PULMONARY ATRESIA WITH VENTRICULAR SEPTAL DEFECT (PA-VSD)

Pulmonary atresia with VSD is a condition characterized by atresia of the pulmonary tract with a perimembranous VSD. This was previously referred to as severe tetralogy of Fallot (TOF).

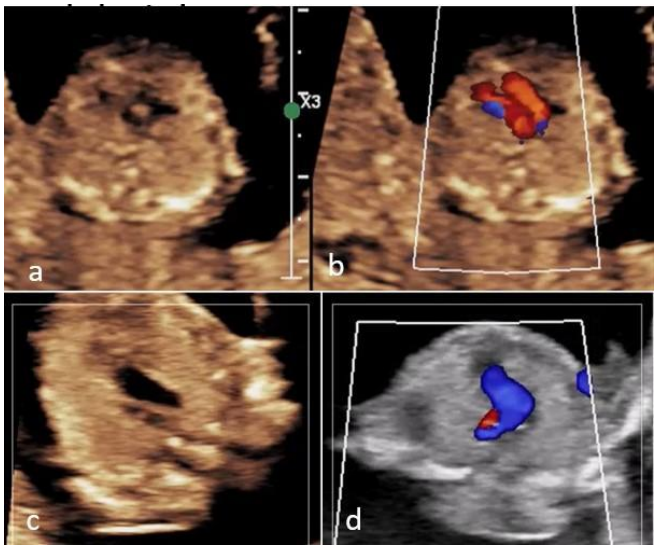


Fig.1: (a & b) Four-chamber view in dual mode shows symmetrical ventricular inflows (b) Three-vessel view shows dilated single vessel, which is the aorta (d) Three-vessel-view on color Doppler shows forward flow in aorta and flow reversal in ductus.

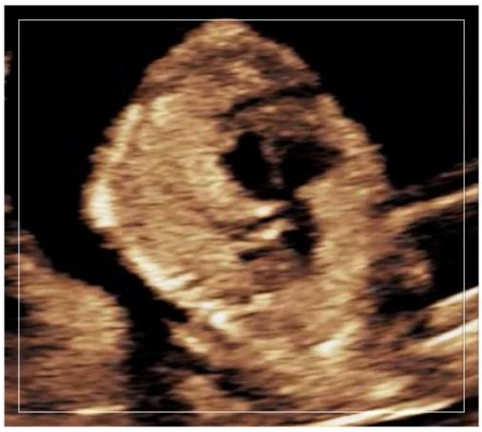


Fig.2: LVOT view shows perimembranous VSD with loss of septo-aortic angle.

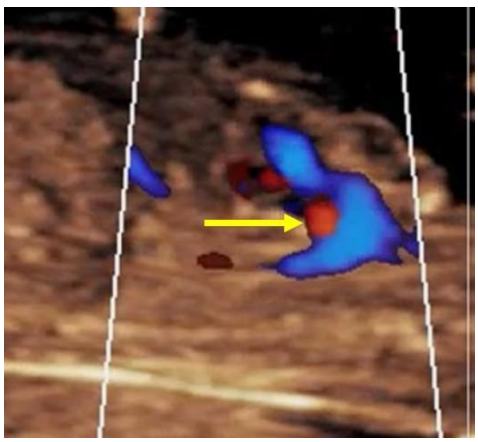


Fig.3: Aortic arch view on color Doppler shows flow reversal in the ductus arteriosus under the arch (arrow).

ULTRASOUND FEATURES

- Four-chamber view is commonly normal in PA-VSD (Figs. 1 a & b).
- Five-chamber view shows VSD with loss of septo-aortic angle (Fig. 2).
- Three-vessel-trachea view shows a single vessel on gray scale. Color Doppler demonstrates reverse flow in the ductus arteriosus and pulmonary artery (Figs.1 c & d).
- Aortic arch view shows reversal of flow in the ductus underneath the arch. This can be seen occasionally in the first trimester (Fig. 3).
- Major aorto-pulmonary collaterals (MAPCAs) are not detected in the first trimester.

DIFFERENTIAL DIAGNOSIS

- Tetralogy of Fallot (TOF) with pulmonary stenosis: pulmonary artery and ductus are narrowed but not atretic. Color Doppler shows forward flow. MAPCAs are not visualised.
- TOF with absent pulmonary valve syndrome: dilated right ventricle in late stages of gestation. Markedly dilated main pulmonary artery. Dilated right and left pulmonary arteries give a butterfly appearance. Color Doppler shows to-and-fro flow in the pulmonary artery. Ductus arteriosus and MAPCAs are usually absent.

ADDITIONAL POINTS

- LVOT view is very important to differentiate PA-VSD from other conditions.
- A right-sided aortic arch is seen in 20% to 50% of cases.
- MAPCAs are associated in about 44% of cases and are typically diagnosed in the second trimester of pregnancy.
- 8.3% of children with PA-VSD have chromosomal anomalies in a study.
- 22q11 microdeletion is seen in 18% to 25% of fetuses with PA-VSD.

TRANSPOSITION OF THE GREAT ARTERIES

Transposition of the great arteries (TGA) is a condition characterized by atrioventricular concordance and ventriculoarterial discordance (complete TGA) or atrioventricular and ventriculoarterial discordance (corrected TGA). The diagnosis of TGA in the first trimester is challenging owing to the small size of the structures.

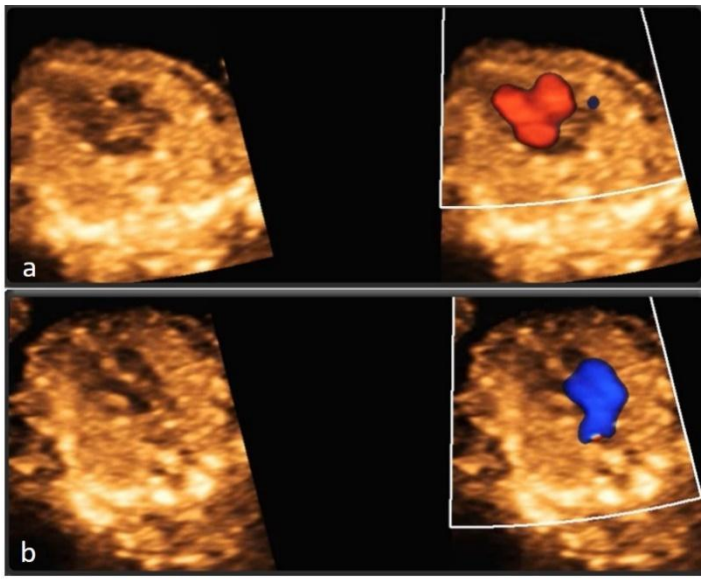


Fig.1: (a) Four-chamber views in grayscale and on color Doppler show normal axis and symmetrical ventricular inflows (b).Outflow tract views show parallel orientation of the outflow tracts.

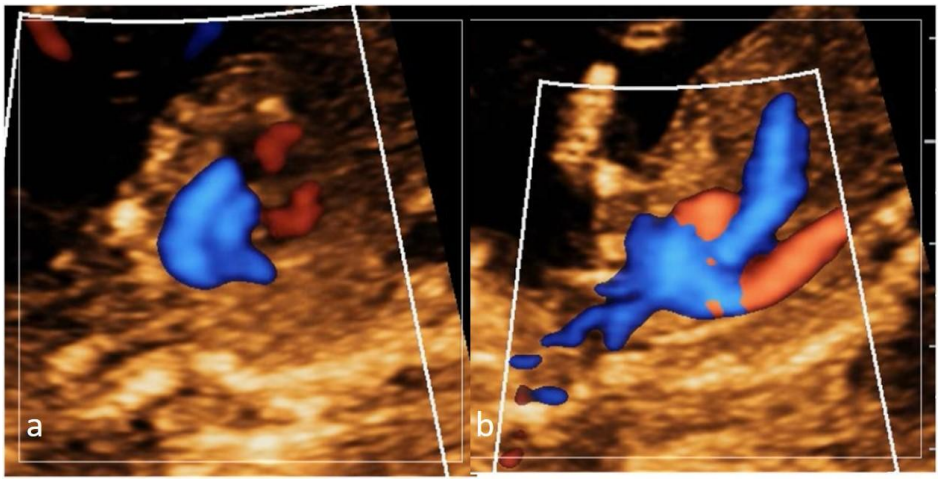


Fig.2: (a) Oblique view shows parallel orientation of the great vessels (b).Arch view shows aorta arising from the anterior-most ventricle, giving branches to the neck.

ULTRASOUND FEATURES

- Four chamber view is usually unremarkable. Symmetrical ventricular inflows are seen (Fig.1a). In some cases, abnormal cardiac axis (mesocardia) is present.
- Three- vessel view shows a single vessel with a convexity (boomerang sign) (Fig. 1b).
- Oblique view is very important, shows the outflow tracts parallel to each other (Fig. 2a)
- Arch view shows the aortic arch arsing from the anterior most ventricle in complete transposition, giving off the branches of the neck vessels (Fig. 2b).

DIFFERENTIAL DIAGNOSIS

- Double outlet right ventricle (DORV): four-chamber view shows altered axis; three-vessel view shows a single vessel and discrepant size of the great arteries; oblique view shows parallel orientation of the great vessels arising from the right ventricle.

ADDITIONAL POINTS

- Oblique view of the fetal chest is not a standard plane of the fetal heart examination. It must be performed when a single vessel is seen on the three-vessel-trachea view.
- Demonstration of a single great vessel on color Doppler in the three-vessel-trachea view is often the first clue for the presence of TGA in the first trimester.
- The diagnosis can be confirmed in the first trimester by demonstrating the parallel orientation of the great vessels in the oblique view.

EBSTEIN ANOMALY

Ebstein anomaly is a condition characterized by inferior displacement of the septal and posterior leaflets from the annulus to the apex of the heart.

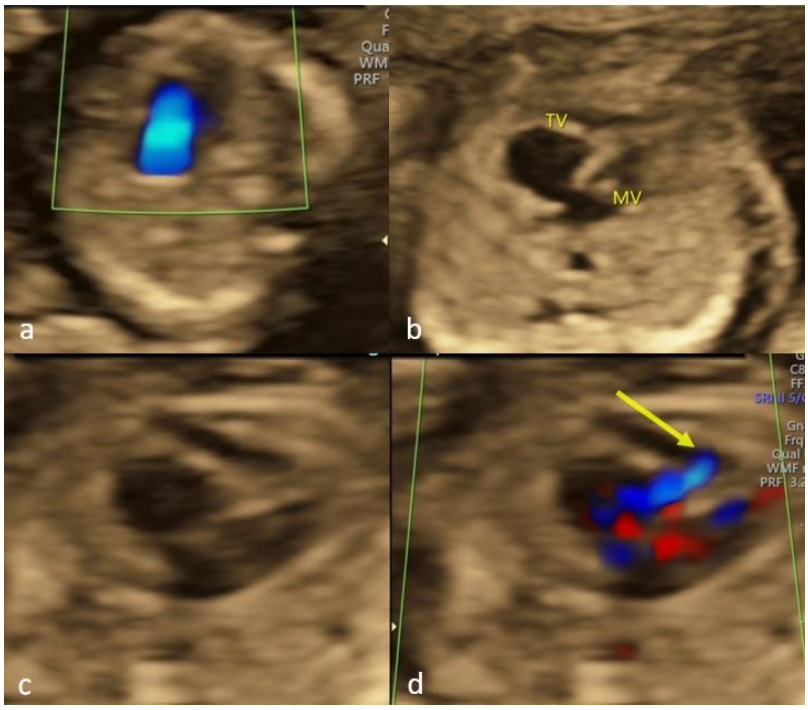


Fig.1: (a) at 12weeks of gestation. Four-chamber view shows significant tricuspid regurgitation. (b) at 14weeks, four-chamber view shows displaced tricuspid valve (c & d) Dual mode examination at 14 weeks shows displaced tricuspid valve with a regurgitant jet arising from deep within the right ventricle (arrow). Patient underwent invasive testing, which revealed trisomy 21.

ULTRASOUND FEATURES

- In the first trimester, a significant tricuspid regurgitation raises suspicion for Ebstein anomaly (Fig. 1a)
- Displaced valve leaflets can be seen on gray scale on an apical four-chamber view (Fig. 1b)
- Color Doppler shows a regurgitating jet arising from deep within the right ventricle, which ricochets against the right atrial wall. (Fig. 1d)

DIFFERENTIAL DIAGNOSIS

- Tricuspid regurgitation: This is a close differential diagnosis in the first trimester. The valve leaflets are in a normal position and are not displaced apically. The regurgitant jet arises at the level of the tricuspid valve annulus (Fig.2).

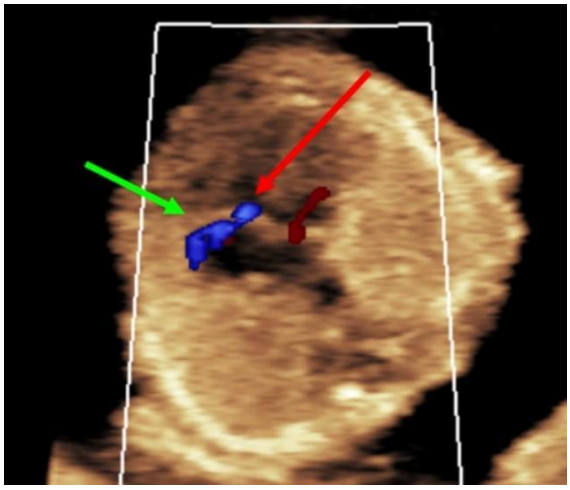


Fig.2: A 13-week pregnancy showing tricuspid regurgitation. The jet originates at the valvular level (red arrow). The valve leaflets (green arrow) are not displaced

ADDITIONAL POINTS

- In cases of severe Ebstein anomaly, presence of cardiomegaly is associated with increased nuchal translucency (NT) and hydrops.
- Cases of mild Ebstein anomaly are missed in the first trimester and are detected in the second or even the third trimester.
- Atrial septal defect is a common association with Ebstein anomaly.
- Most of the cases are isolated, but association with trisomy 21 and trisomy 13 has been reported.

TETRALOGY OF FALLOT

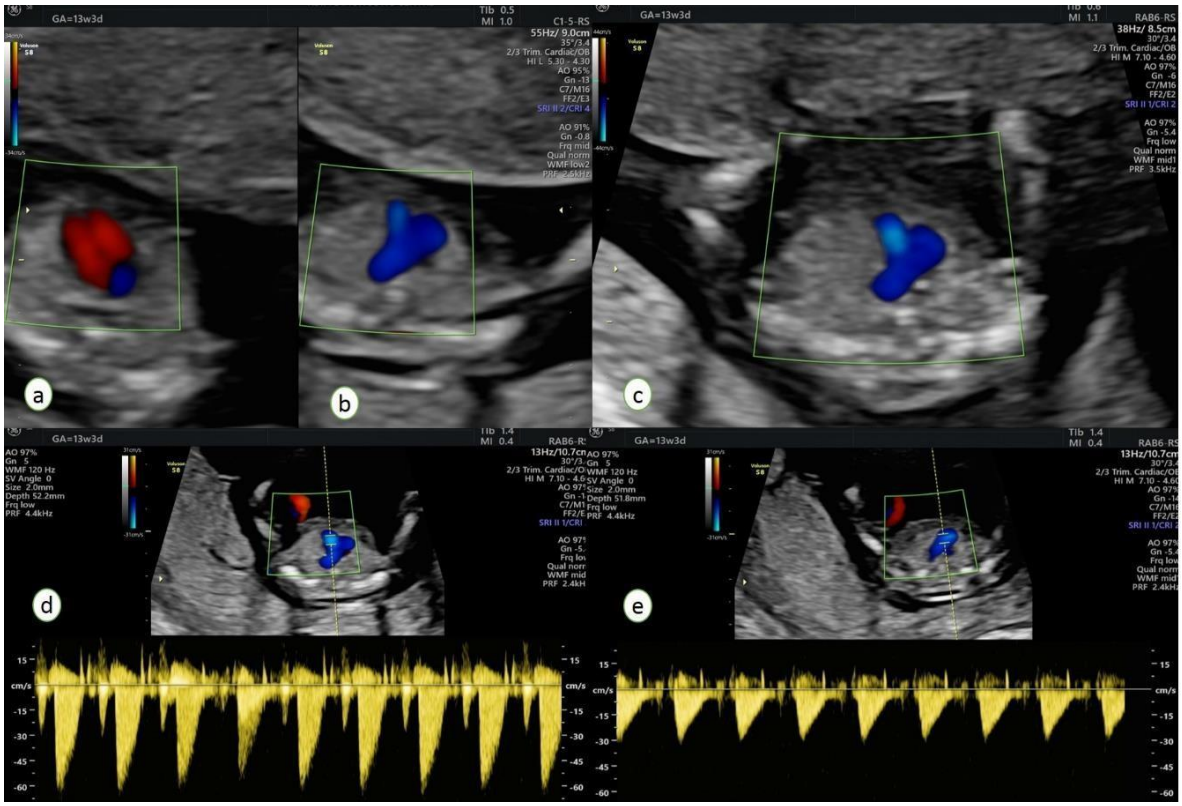


Fig.1: Tetralogy of Fallot in a 13-weeks fetus (a) Normal four-chamber view (b) Three-vessel-trachea view shows a narrow-caliber RVOT forming a 'Y' sign (c) High velocity flow with aliasing is seen in the RVOT (d) High velocity antegrade flow noted in the RVOT on spectral Doppler (e) Normal velocity antegrade flow seen in the LVOT. ***Case contributed by Dr. Nitin Jadhav***

ULTRASOUND FEATURES

- Four-chamber view is normal.
- The pulmonary artery is narrow in caliber and forms a "Y" sign with the aorta.
- Antegrade flow is noted in both outflow tracts. Aliasing is noted in the RVOT across the pulmonary valve.
- Perimembranous VSD and overriding of the aorta are noted in all cases at later stages.

DIFFERENTIAL DIAGNOSIS

- Pulmonary atresia with VSD
- Primary pulmonary valve dysplasia/stenosis
- Common arterial trunk

ABSENT PULMONARY VALVE

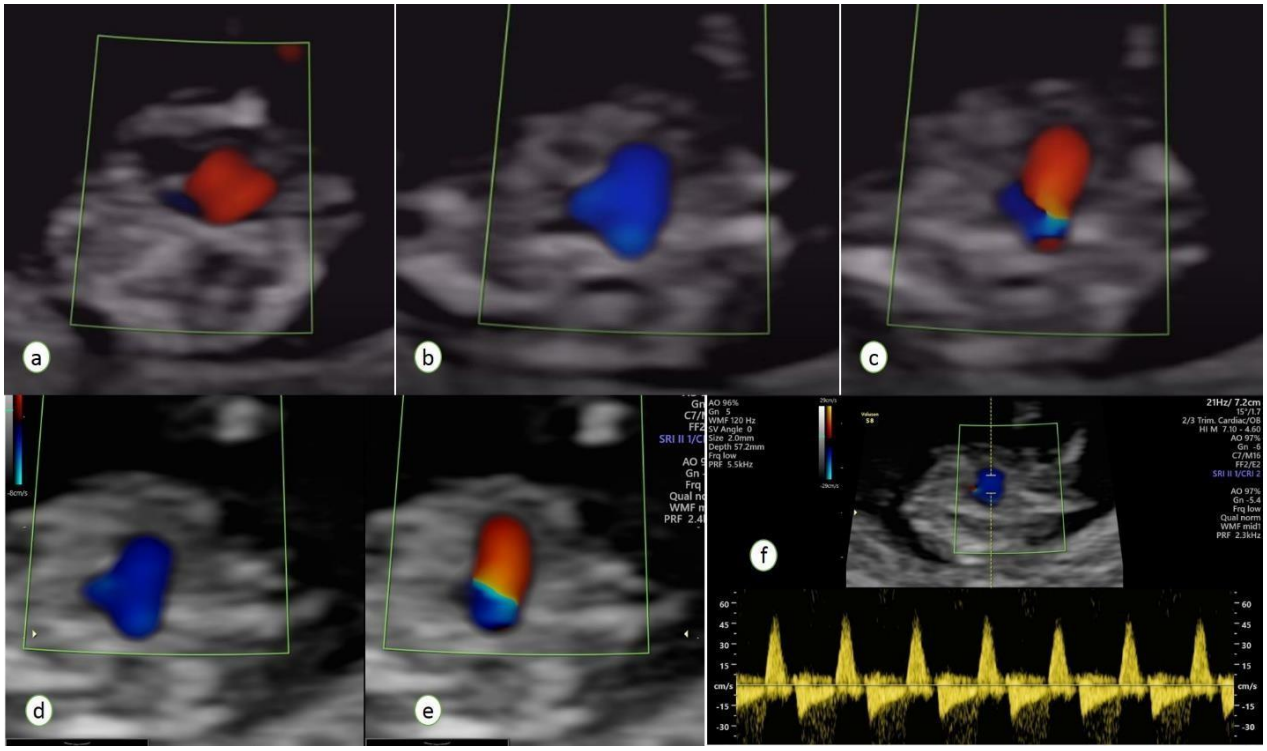


Fig. 1: Absent pulmonary valve in an 11-weeks fetus (a) Normal inflow tracts (b) Both outflow tracts showing antegrade flow (c) RVOT showing reversal of flow direction (d and e) Bidirectional flow in the RVOT (f) Bidirectional flow on Spectral Doppler. ***Case contributed by Dr. Nitin Jadhav***

ULTRASOUND FEATURES

- Four-chamber view is normal in early scans.
- The outflow tracts are also of normal caliber in the early scans.
- A typical bidirectional flow is an obvious finding on color Doppler and spectral Doppler.
- During the later stages of gestation, dilatation of the main pulmonary artery and its branches becomes more evident.

DIFFERENTIAL DIAGNOSIS

- Tetralogy of Fallot
- Primary pulmonary valve dysplasia/stenosis
- Common arterial trunk

ABSENT SEMILUNAR VALVES VALVES

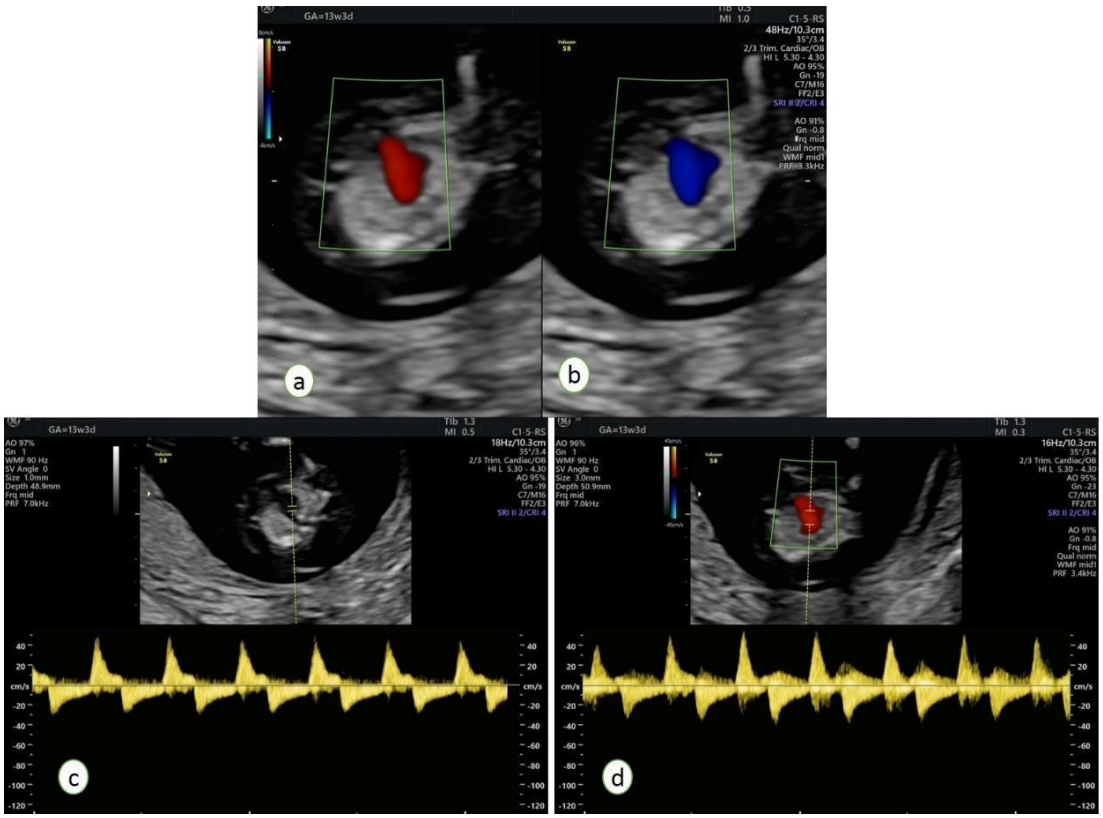


Fig.1: Absent semilunar valves in a 13-weeks fetus (a) Retrograde flow in the outflow tracts in diastole (b) Antegrade flow in the outflow tracts in systole (c) LVOT showing bidirectional flow on spectral Doppler (d) RVOT showing bidirectional flow on spectral Doppler. ***Case contributed by Dr. Nitin Jadhav***

ULTRASOUND FEATURES

- Four-chamber view is normal in early scans.
- The outflow tracts are also of normal caliber in the early scans.
- A typical bidirectional flow is an obvious finding on color Doppler and spectral Doppler.
- Bidirectional flow across the pulmonary and aortic valve favors valvular pathology like agenesis or grossly dysplastic valves

CORONARY ARTERY FISTULA

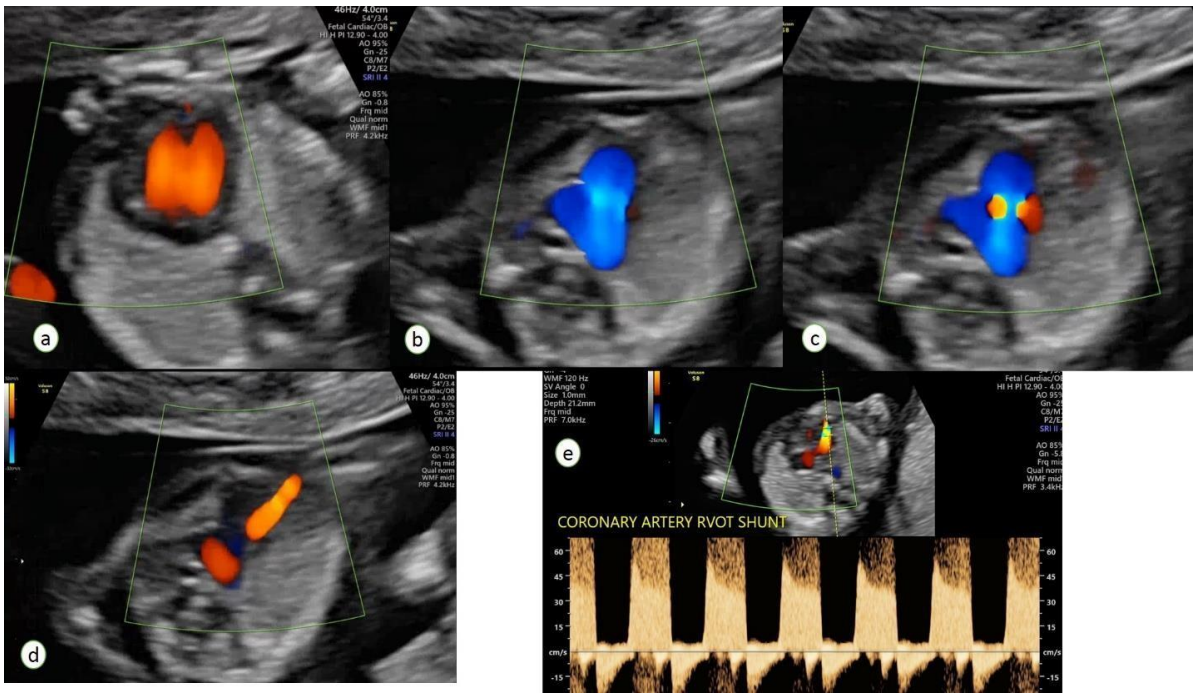


Fig.1: Coronary artery fistula in a 12-weeks fetus (a) Normal four-chamber view of the heart (b) Both outflow tracts showing normal antegrade flow (c) RVOT showing aliasing at the insertion site of the fistula (d) High-flow coronary artery fistula extending from the cardiac apex up to the RVOT (f) Spectral Doppler showing high velocity flow in the fistula. ***Case contributed by Dr. Nitin Jadhav***

ULTRASOUND FEATURES

- Early detection of coronary artery fistula at 12 wks is extremely rare.
- Four-chamber view is normal.
- The outflow tracts are also of normal caliber and show antegrade flow.
- In this case, the coronary artery fistula is seen extending from the cardiac apex and seen traversing cranially, to open into RVOT near its bifurcation.
- The fistula shows aliasing on color Doppler and high velocity flow on spectral Doppler.
- Many types of coronary artery fistulas are reported in the postnatal series with their communications with different chambers of the heart as well as with outflow tracts.
- These fistulas may be associated with other abnormalities. This case was associated with lower limb abnormalities and situs ambiguous.

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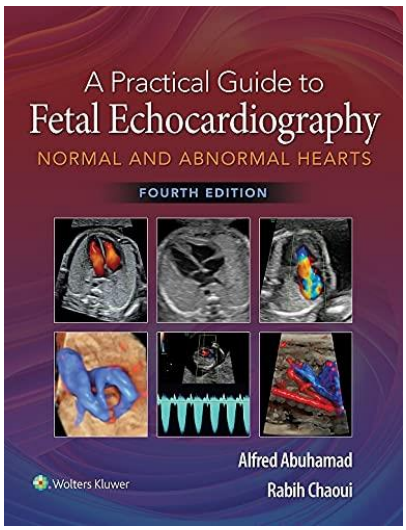

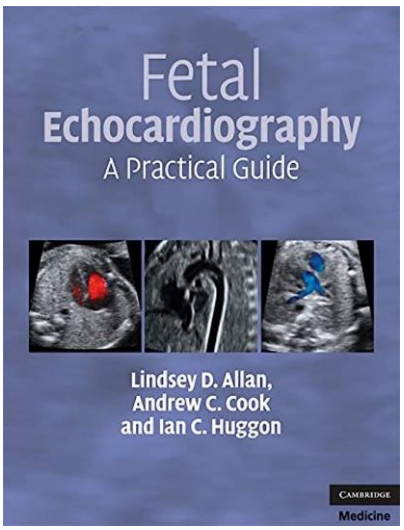

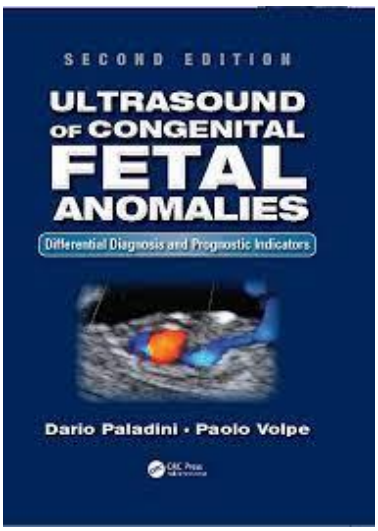

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