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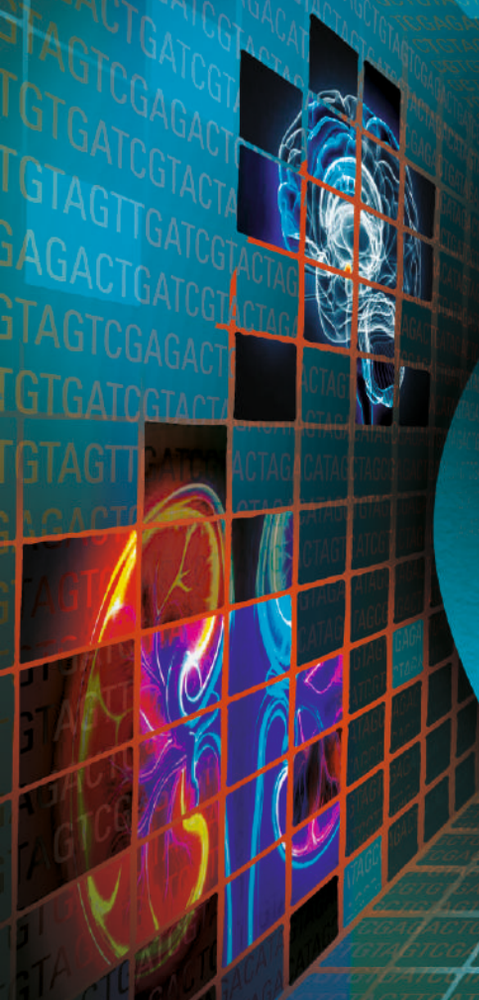
SOCIETY OF FETAL MEDICINE

Society of Fetal Medicine
Bengal Chronicles

BeatWise

The Cardiac Lingo



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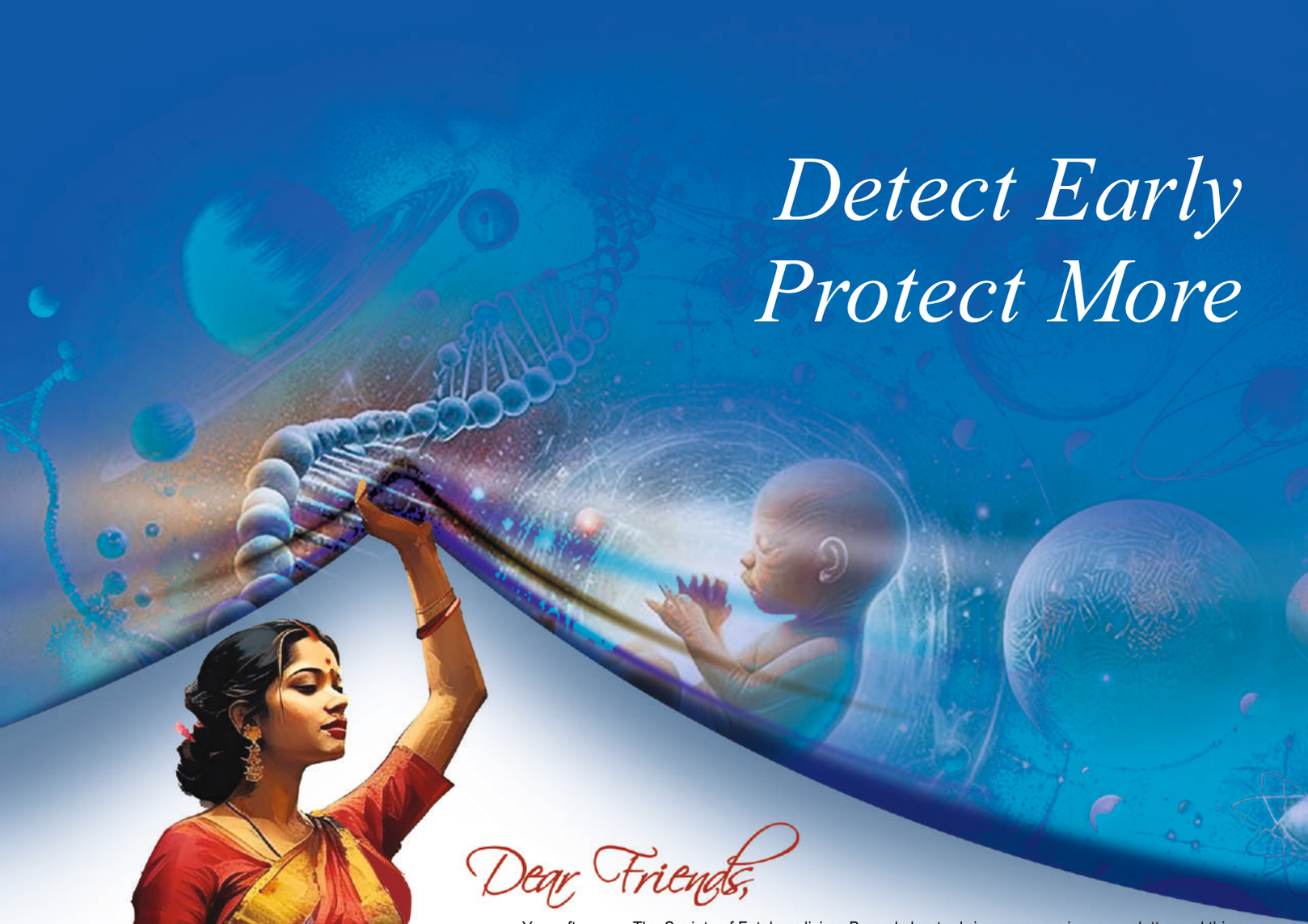
Detect Early Protect More



Dear Friends,

Year after year, The Society of Fetal medicine, Bengal chapter brings you a unique newsletter and this year too we are back with yet another interesting one.
It gives me great pleasure to present Beatwise, a newsletter dedicated to the ultrasound evaluation of the fetal heart and the diagnosis of cardiac abnormalities in the prenatal period. The Fetal heart, with its intricate anatomy and dynamic physiology, represents one of the most fascinating yet challenging areas in prenatal imaging.

Over the past decade, remarkable advances in ultrasound technology, improved imaging protocols, and enhanced training have significantly strengthened our ability to evaluate the fetal heart. What was once limited to basic screening has now evolved into comprehensive fetal cardiac assessment, enabling early detection of congenital heart disease and more informed clinical decision-making.

Early identification of cardiac anomalies during pregnancy plays a crucial role in optimizing outcomes. Accurate prenatal diagnosis allows timely referral for fetal echocardiography, appropriate counseling for families, coordinated perinatal care, and when necessary, delivery planning at specialized centers with neonatal cardiac support. Such multidisciplinary collaboration between obstetricians, fetal medicine specialists, radiologists, and pediatric cardiologists is key to improving neonatal survival and long-term outcomes.

Beatwise has been envisioned as a platform to promote learning, share clinical experience, and encourage best practices in fetal cardiac screening. Through expert contributions, case discussions, imaging insights, and updates on evolving protocols, this newsletter aims to strengthen confidence and competence in evaluating the fetal heart across practitioners involved in prenatal care.

We are immensely grateful, as always , to our inspiring forces, our Mentor Emiratus, Professor Ashok Khurana, our National President Dr Mohit Shah and the entire central team for giving the Bengal chapter the opportunity and entrusting us with this responsibility. As we continue to advance in this field, it is important to remember that every scan represents not just a diagnosis but a family seeking reassurance and guidance. Our commitment to accuracy, compassion, and continuous learning remains central to our work.

I congratulate the editorial team, especially Dr Kanchan Mukherjee and Dipanjana Datta and the contributors for their efforts in bringing this initiative to life and look forward to Beatwise becoming a valuable resource for the Fetal medicine community. Let 'Beatwise' be the rhythm that keeps our curiosity alive and our hearts beating for excellence in Fetal cardiac imaging.

Happy Reading and Happy Relearning!
Long Live SFM
Jai Hind

Dr Seetha Ramamurthy Pal
President, Society of Fetal Medicine, Bengal Chapter

Foreword

Dear Colleagues,

Few moments in medicine are as profound as watching a tiny heart beating inside the womb. In those moving images lies not only science and technology, but also hope, responsibility, and the promise of a new life.

Fetal cardiac imaging has advanced tremendously in recent years, allowing radiologists and fetal medicine specialists to detect and understand congenital heart disease with increasing accuracy. Equally important is the collaborative effort between radiologists, obstetricians, pediatric cardiologists, and neonatologists that helps translate these diagnoses into better care and outcomes.

This issue of Beatwise celebrates that spirit of learning and collaboration. I congratulate the editorial team and contributors for their dedication in bringing together knowledge, experience, and insight for the benefit of our professional community.

May this publication continue to encourage curiosity, dialogue, and progress in fetal imaging.

Dr Mohit Shah
President, Society of Fetal Medicine

SCREEN SMART: UPDATED CARDIAC VIEWS EVERY ANOMALY SCAN MUST CAPTURE

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SOCIETY OF FETAL MEDICINE

Congenital heart disease (CHD) is the most common congenital malformations with an incidence of about 8:1000 live births. Though the incidence is even higher in fetal population. A good number of fetuses with complex cardiac anomalies succumb in the first trimester itself, even before the cardiac anomaly is suspected; some parents opt for termination of pregnancy after the diagnosis is made in the mid-trimester; and some cardiac anomalies are progressive and end in intrauterine death. Thus the incidence quoted may only be the tip of the iceberg.

Effective cardiac screening should maximise detection of structural anomalies and abnormalities of function and rhythm, as a part of routine prenatal care. CHD often occurs in low-risk pregnant patients, which underscores the importance of routine cardiac screening at the time of second trimester ultrasound. Prenatal diagnosis of CHD is important for counselling and decision-making, focussed diagnostic testing, and optimal prenatal and delivery management. As a result, prenatal diagnosis has led to improved neonatal and infant outcomes.

GESTATIONAL AGE

The cardiac screening examination is performed optimally between 18 - 22 weeks gestation.

TECHNICAL FACTORS

Ultrasound transducer: Higher frequency probes improves the likelihood of detecting subtle defects, at the expense of reduced acoustic penetration.

Imaging parameters: Cross-sectional greyscale imaging is the basis of a reliable fetal cardiac scan. System settings should emphasise a high frame rate, with increased contrast and high resolution.

Zoom, cine-loop and image storage: Images should be magnified until the heart fills atleast one-third to one-half of the screen. Cine loop feature should be used to assist the real-time evaluation of normal cardiac structures. The examination should be recorded in a manner that will allow subsequent review to verify its diagnostic adequacy with proper patient identification and labelling of image laterality and orientation.

Cardiac examination

The cardiac screening should include the fetal situs and the four-chamber, outflow-tract and great-vessel views.

Situs and the four-chamber view.

To assess cardiac situs, it is necessary first to determine fetal laterality, i.e to identify fetal right and left sides, based on the position of the fetus in utero. In second trimester, the heart is positioned in a horizontal plane within the chest, held in place by the fetal liver, which extends to the left side of the fetal abdominal wall. Cardiac axis or apex points to left by 45 ± 20 degrees. A transverse sweep with cephalad movement of the transducer, from fetal abdomen towards fetal chest, allows visualisation of abdominal situs and four-chamber view.

The abdominal situs is obtained at the level of the standard abdominal circumference measurement, with stomach visible on the left side. Additionally, cross-sectional views of the descending aorta and inferior vena cava are seen on left and right side of the spine, respectively.

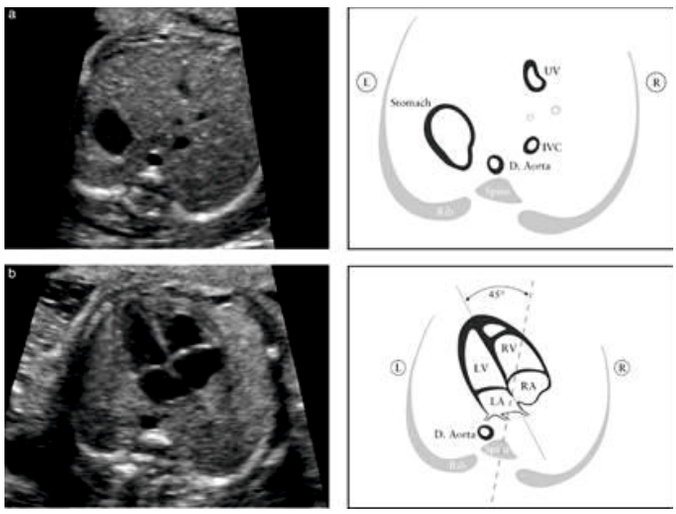

Fig: Schematic diagram and corresponding grayscale images. Abdominal situs ascertained in transverse view of fetal abdomen. Cardiac position and axis with apex pointing to left at an angle of 45±20 degree in relation to antero-posterior axis of chest.

Four-chamber view

Assessment of four-chamber view involves careful evaluation of specific criteria. The main elements for examination of four-chamber are:

Atrial chambers	Ventricular Chambers	Atrio-ventricular junction and valves.
Two atria, approximately equal in size.	Two ventricles, approximately equal in size.	Intact cardiac crux
Foramen ovale flap in left atrium	No ventricular wall hypertrophy	Differential offsetting: tricuspid valve leaflet inserts on ventricular septum closer to cardiac apex than does mitral valve.
Atrial septum primum present (near crux)	Moderator band at right ventricular apex	
Atleast one pulmonary vein entering left atrium.	Ventricular septum intact.(apex to crux)	

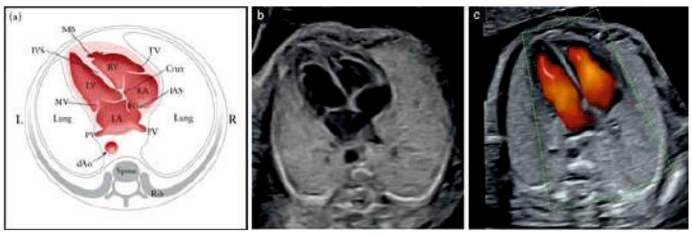

Fig: schematic drawing and corresponding grayscale and color doppler images of four-chamber view

A normal heart is usually no larger than one-third of the area of the chest. A small amount of pericardial fluid (≤ 2mm thickness at end diastole) is commonly seen during the second and third trimesters and is a normal finding.

Normal heart rate and regular rhythm should be confirmed. The normal rate ranges from 120- 160 beats per min. Bradycardia , often associated with transducer pressure on abdomen, is observed transiently in normal second trimester. However, persistent bradycardia in a well fetus requires timely evaluation of the possible causes.

Mild transient tachycardia (160-180 bpm) can occur as a normal variant during fetal movement. Again, persistent tachycardia (>180bpm) should be evaluated further.

Outflow tract view

Sweep technique: Performing a transverse sweep with cephalad movement of the transducer from the four-chamber view towards the upper chest, accompanied by small adjustments in insonation angle, starting from a four-chamber view, to visualise normal cross-over of main pulmonary artery and aorta at its origin, details of pulmonary artery bifurcation.

Rotational technique: This starts from a four-chamber view of heart, with transducer being rotated towards fetal right shoulder. Optimises visualisation of LVOT, especially the outlet part of septum that is in continuity with anterior wall of aorta.

With both the techniques, once LVOT view is obtained, transducer is angled cephalad and rotated towards left fetal shoulder to obtain RVOT view with pulmonary artery is and its bifurcation.

Left ventricular outflow tract (LVOT) view

The LVOT view confirms the presence of a great vessel originating from the morphological left ventricle and from the centre of the heart. Continuity should be documented between the ventricular septum and the anterior wall of this vessel to demonstrate the integrity of the outlet septum. However, it is only the presence of the head and neck vessels originating from it that confirms this vessel as aorta. The aortic valve should move freely and should not be thickened.
Sagittal views of aortic and ductal arches and assessment of the neck vessels are currently not considered part of routine cardiac screening examination.

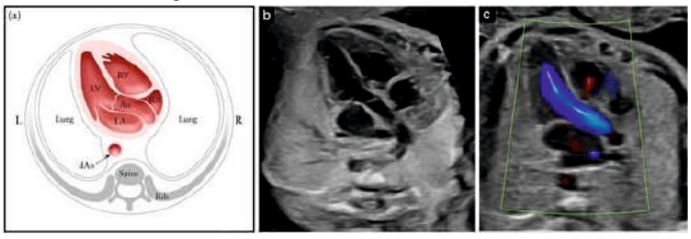

Fig: Schematic diagram and corresponding grayscale and color Doppler images of left ventricular outflow tract view imaged from apical approach.

Right ventricular outflow tract (RVOT) view

The RVOT view confirms the presence of a great vessel, the pulmonary artery, originating from the morphological right ventricle and branching after a short course. The pulmonary valve should move freely and should not be thickened.
Pulmonary artery is usually slightly larger than the ascending aorta during fetal life and crosses the ascending aorta anterior and cephalad to LVOT at almost right angle. It courses towards the left of most posterior ascending aorta, which is seen in cross-section.At this level, superior vena cava is seen right of the aorta.

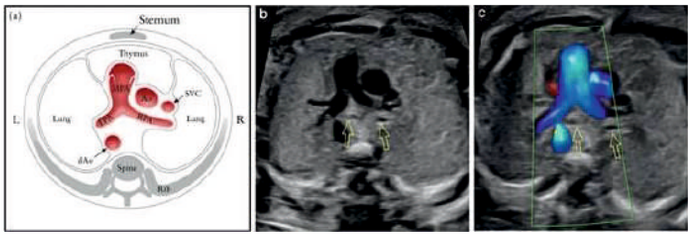

Fig: Schematic diagram and corresponding grayscale and color Doppler images of right ventricular outflow tract view, which relates closely to three-vessel view.

Three-vessel (3VV) view and three-vessel-and-trachea (3VTV) view

Three vessel view evaluates the pulmonary artery, ascending aorta and superior vena cava; their relative sizes , relationships and also assessment of vessel number. From left to right, the vessels are pulmonary artery, aorta and right superior vena cava. Their relative diameters decrease from left to right.
Three vessel trachea view is a view cephalad with respect to the 3VV, in which the transverse aortic arch is visualised and its relationship with the trachea emphasized.This view shows the main pulmonary artery in direct communication with the ductus arteriosus. The normal transverse aortic arch is positioned to the right of ductal arch. Trachea can be identified as a hyperechogenic ring surrounding a small fluid filled space. The normal ductus arteriosus and aortic arch course to the left of trachea and form an acute angle (V- shaped). Right superior vena cava and thymus is also seen in this plane.

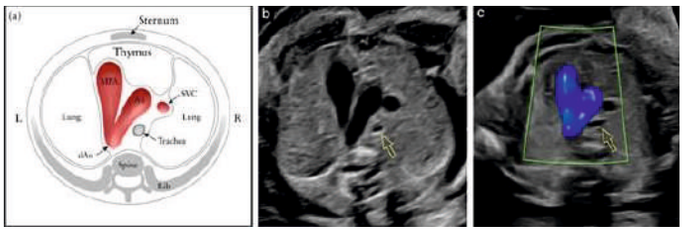

Fig: Schematic diagram and corresponding grayscale and color Doppler images of three-vessel-and-trachea view.

Color flow Doppler Ultrasound in Cardiac Screening

Color flow mapping is an integral part of fetal cardiac screening .It demonstrates antegrade flow across the atrio-ventricular valves, semilunar valves and great arteries. It helps to highlight abnormal blood flow patterns, such as shunts, AV valve regurgitation and flow reversal in ductus arteriosus. It helps to evaluate cardiac anatomy in obese patients.

Early fetal cardiac screening

Transvaginal transducers or high-frequency transabdominal transducers may be used. Color and power doppler ultrasound should be adjusted to prioritize the color signal over greyscale, to enhance visualisation of blood flow across the small structures in first trimester fetal heart.

Optimum timing of cardiac screening in first trimester : 12+3 weeks till 13+6 weeks.

Components of a detailed early cardiac screening examination are as follows:

1. Situs: on greyscale imaging to ascertain normal position of the stomach and heart, both of which should be on the left side of fetus.

2. Four-chamber view: displayed using both grayscale and color/ power doppler with demonstration of biventricular filling.

3. Three vessel trachea view : displayed using color/ power doppler, demonstrates left sided aortic and ductal arches.

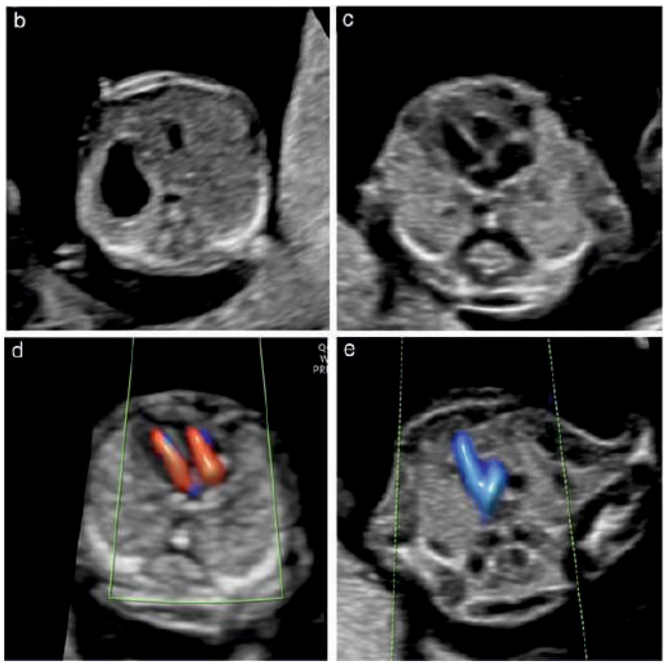

Fig: Cardiac examination in early gestation should focus on these selected planes: demonstration of cardiac axis and abdominal situs with stomach on left side, four-chamber view (color doppler) during diastole and three-vessel-and- trachea view (color doppler) during systole.

Common pitfalls of cardiac screening examination

1. Missed abnormalities: small ventricular septal defects (VSDs), mild valve abnormalities are frequently missed.
2. Drop-out artifact: A normal four-chamber view may be misdiagnosed as a VSD when the beam is parallel to the septum.
3. Optimal timing: screening too early (before 18 weeks) may miss abnormalities that become apparent later, while scanning in third trimester can be limited by acoustic shadowing.
4. Lesion progression and timing: Some lesions such as coarctation of aorta or pulmonary stenosis, may not be apparent in early pregnancy and can develop or worsen later.
5. Technical factors: Maternal obesity, reduced amniotic fluid and unfavourable fetal position (prone) significantly reduce image quality, leading to poor visualisation of cardiac structures.

Conclusion

Fetuses identified as having or suspected of having an abnormality on routine cardiac ultrasound screening are candidates for a fetal echocardiogram. For fetuses with a significant risk factor for CHD, i.e. when their risk is elevated above that of general population , fetal echocardiography is also indicated in addition to routine cardiac screening. However, a high proportion of cases with a CHD detectable prenatally are patients without any risk factors or extracardiac anomalies, hence the importance of quality screening, with timely referral if this suggests an abnormality.

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From Four- Chamber To Outflows: A Stepwise Echo Workflow

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Congenital heart disease (CHD) remains the most common congenital anomaly worldwide. Despite advances in ultrasound technology, a significant proportion of critical CHDs are still missed during routine antenatal screening. The reason is rarely lack of equipment — it is usually the lack of correct method. A structured, sequential, and reproducible workflow is the difference between pattern recognition and true cardiac screening.

This article outlines a practical echocardiography workflow — moving deliberately from four-chamber assessment to outflow tract evaluation — ensuring that no CHD is overlooked. With this, we should be able to answer three fundamental questions:

1. Are the cardiac segments morphologically normal?
2. Are they correctly connected?
3. Are they spatially related as expected?

With these questions borne in mind, we move ahead with the following workflow.

1. Assessing the fetal laterality and abdominal situs: The workflow begins not looking first at the heart, but in abdomen. The normal arrangement includes: stomach and spleen on left, majority of liver on right, aorta to left of spine and IVC to right and anterior to aorta.

2. Cardiac situs: The heart should normally be situated on the left, with its apex pointing towards the left. Cardiac axis should be noted (normal 45 +/- 20)

Any abnormality in cardiac position (mesocardia, dextrocardia) or axis deviation or translational shift (dextroposition) should be looked for.

These two views are important to determine situs solitus (normal arrangement) and its abnormality raises suspicion of situs inversus or ambiguous.

3. 4 chamber view: It is the basic view for cardiac screening.

• AV connection: First we have to identify the 4 chambers based on its anatomical features and not solely on spatial orientation. The morphological right atrium has broad based triangular appendage, morphological left atrium has left atrial appendage with narrow base and is tubular. This helps to look for any atrial isomerism. The morphological right ventricle has a moderator band and a tricuspid valve with slightly lower insertion on interventricular septum (IVS) as compared to mitral valve. The morphological left ventricle has smooth walls with fine apical trabeculations. Identifying these helps in understanding the correct atrioventricular connection, thus reducing chances of missed CHDs like corrected TGA.

• **Cardiothoracic ratio:** normal (0.5-0.6) to rule out cardiomegaly

• **Cardiac crux:** It is crucial to view the lower atrial septum meeting the upper ventricular septum at the insertion of AV valves. We should be able to see the crux properly in a normal heart, if we are not too cranial or too caudal. Normal offsetting should be mandatorily looked for, to miss possible AVSD.

• **Interatrial septum and patent foramen ovale:** We should look for normal flow from right to left across foramen ovale. Atrial septal defect and atrial septal aneurysm (though rare) can be picked up.

• **Intervertricular septum:** The intactness of the IVS should be looked for, both in apical/ basal and lateral views to rule out any inlet, muscular and some perimembrabous VSD. Mid-muscular VSDs are best picked up on lateral views.

• **Chamber symmetry:** This helps in ruling out any hypoplastic ventricle, Ebstein anomaly, malalignment VSD, possible coarctation of aorta etc.

• **AV valves:** It is important to note the normal opening and closing of the valves in cardiac cycle, rule out any possible atresia, stenosis or thickening. If any abnormality is noted, PSV across the valve might help in differentiating these conditions better.

• **Rhythm :** Normal sinus rhythm should be documented, and if any abnormality is noted, we must proceed to look for any possible AV conduction defects.

• **Contractility:** Normal contractility should be looked for, to miss any cardiomyopathy or endocardial fibroelatosis.

• **Venoatrial connection:** Last but not the least in 4 chamber view, it is mandatory to look for at least two pulmonary veins opening into the left ventricle (best confirmed by pulse doppler insonation) and “the space behind the heart”, i.e., the distance between the posterior part of left atrium and descending aorta should not be increased, in which cases TAPVC or PAPVC shoule be suspected.

• **Coronary sinus:** It is normally visualised when we are caudal from 4 chamber view. However, dilated coronary sinus might indicate PLSVC or cardiac TAPVC.

4. Outflow tract views: Cardiac examination is never complete just at the 4 chamber view. Next, we should carefully examine the outflow tracts. LVOT is obtained by slightly angulating the probe, while sweeping up from 4Ch view. RVOT is seen by rotating the probe on opposite side and higher than LVOT.

• **Ventriculoarterial connection:** We should look for aorta arising from morphological left ventricle (which does not bifurcate and gives rise to head and neck branches in upper mediastinum) and pulmonary artery (PA) arising from morphological right ventricles (which bifurcates into two branches in chest, soon after its origin). It is always a good practice to document the "crossover". Conotruncal anomalies like Tetralogy of Fallot (TOF), Double outlet right ventricle (DORV) and Transposition of great arteries (TGA) can be picked up with these views.

• **Septo-aortic continuity:** Anterior wall of aorta should be seen as continuous with IVS and posterior wall with mitral valve. Any abnormality here along with bidirectional colour flow raises suspicion of perimembranous VSD, and when it is associated with aorta receiving blood from both ventricles, we have a diagnosis of aortic override.

• **Semilunar valves:** Both aortic and pulmonary valve should be seen moving freely and disappearing during systole, which rules out any stenosis or atresia. If any abnormality is noted, spectral doppler can be used to differentiate between the conditions.

5. Three vessel view (3VV) and 3 vessel trachea view (3VT): Next, we sweep more cephalad from apical 4ch view. We should be able to see PA, aorta and superior vena cava (SVC) from left to right, in descending order of their sizes in 3VV. The Ductus arteriosus and aortic arch are seen to form a "V" shaped configuration on left of trachea at 3VT view.

• **Number of vessels:** Normally, we should see only 3 vessels in 3VV. If there are four vessels, this raises suspicion of persistent left superior venacava (PLSVC) or a vertical vein of supracardiac TAPVC, which is differentiated by direction of colour flow (same direction as other vessels in PLSVC, opposite direction to SVC in supracardiac TAPVC) and presence / absence of left brachiocephalic vein on a higher view. If there are 2 vessels (one great artery and one SVC) we should suspect conotruncal anomalies like TOF, DORV, CAT.

• **Proportion of size of vessels:** If any disproportion of relative sizes are noted in 3VV, anomalies like pulmonary stenosis/ atresia (where PA< Aorta) ; aortic stenosis/ atresia, coarctation of aorta, tubular hypoplasia, interrupted aortic arch (where PA>> Aorta) should be considered. PA/ Ao ratio >1.6 may be indicative of coarctation of aorta.

• **Ductus arteriosus and transverse aortic arch:** The ratio of Ductus arteriosus to aortic isthmus can be measured at the level of trachea (normal >1) . Concordant flow (same colour direction in both vessels) should be noted here. Discordant flow or turbulence might indicate valve atresia, or severe coarctation of aorta. Any deviation from the normal "V" shape may indicate right aortic arch (U shaped configuration around trachea) or double aortic arch (O shape). Thymus box is visualised at this level, anterior to 3VT.

6. Upper mediastinal view: Cardiac assessment should be further continued with sweeping up from 3VT, which allows visualisation of "S" shaped vessel, the Right subclavian artery, normally present in front of trachea. Any vessel posterior to trachea in this view should alert us about Aberrant Right subclavian artery (ARSA), which should be differentiated from left brachiocephalic vein by insonating with pulse Doppler.

7. Longitudinal views: Cardiac evaluation should include the longitudinal arch views and bicaval view.

• Aortic arch: Aortic isthmus must be checked along with colour flow. Any narrowing of aortic isthmus (Z score <-2SD or shelf sign) with any note of turbulence should raise suspicion of coarctation. The proper "candy cane" appearance should be seen to rule out any narrowing of a longer segment (tubular hypoplasia) or interrupted segment near neck vessels (interrupted aortic arch).

• Bicaval view: SVC and IVC should be eyeballed for a normal appearance. Any dilatation of SVC may indicate TAPVC.

8. Aorta and IVC: Lastly, the continuity of the great vessels including aorta and IVC should be checked. Any interruption of IVC might indicate heterotaxy, or an extra vessel between aorta and IVC crossing the diaphragm may indicate infracardiac TAPVC.

As per Carvalho's key teaching, complex CHDs become simple when described segment by segment. Some CHDs can wait few weeks to months postnatally to get detected and be treated, while some lives are lost in early neonatal life just because of lack of a thorough examination of fetal heart in prenatal period. Advances in technology have helped us delineate the structures of heart right from first trimester, and we should utilise every opportunity we get to screen the heart once more before a life is born. It is rather important to approach all the segments methodically one by one, reduce the chances of missing any cardiac disease to the best of our ability and prepare the couple for a smooth transition from prenatal to neonatal period.

Dive Deeper

SOCIETY OF FETAL MEDICINE

Beyond 2D: Unlocking 3D/4D & STIC for Spatial Cardiac Clarity

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SOCIETY OF FETAL MEDICINE

Congenital heart disease (CHD) remains the most common congenital anomaly and a leading contributor to perinatal morbidity and mortality worldwide. Despite major advances in prenatal screening, conventional two-dimensional (2D) fetal echocardiography continues to be highly operator dependent and sometimes limited in depicting complex spatial relationships. The advent of three-dimensional (3D) and four-dimensional (4D) ultrasound—particularly Spatio-Temporal Image Correlation (STIC)—has transformed fetal cardiac imaging by enabling volumetric acquisition, offline analysis, and improved reproducibility.

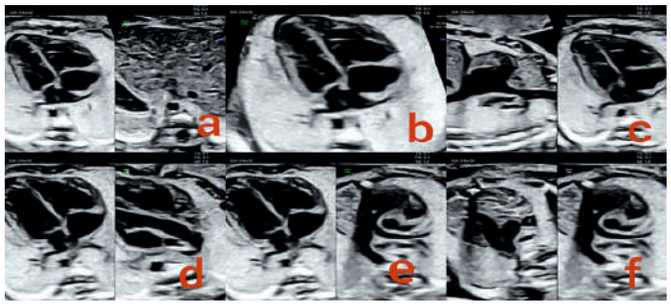

Figure 1. 2D gray scale cardiac planes from upper abdomen, situs(a); four chamber (b);pulmonary veins and systemic venous connections (c);left ventricular outflow tract (d);right ventricular outflow tract (e);and three vessel trachea view (f)

3D/4D imaging acquires complete volume datasets rather than isolated 2D slices, thereby reducing operator dependency and allowing reproducible offline review. These advantages are particularly valuable in busy obstetric practice and in settings where expert fetal cardiology support is limited.

Principle of STIC

At the core of modern 3D/4D fetal cardiac assessment lies Spatio-Temporal Image Correlation (STIC). STIC combines spatial information obtained through a motorized 3D sweep with temporal information from the fetal cardiac cycle. The system reconstructs a dynamic cine loop representing the beating fetal heart. This gated reconstruction allows the examiner to review cardiac motion repeatedly and in multiple planes.

The STIC acquisition process requires attention to technical details such as correct probe positioning, ensuring fetal quiescence, selecting an appropriate sweep angle (typically 25–45 degrees), and encouraging brief maternal breath holding to reduce motion artifacts. When optimized, the technique yields a robust volumetric dataset suitable for detailed offline interrogation.

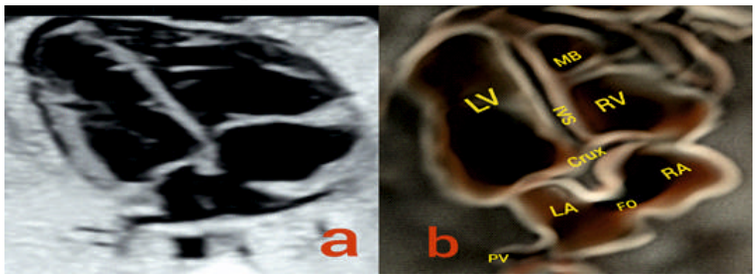

Figure 2.2D gray scale 4 chamber view (a), and STIC rendered 4 chamber view of fetal heart (b)

Optimization and Technical Considerations

High-quality datasets depend heavily on acquisition parameters. Optimization of acquisition requires the need to balance gain to avoid noise or dropout, maintenance of adequate frame rate for smooth temporal resolution, and selection of an appropriate sweep time—usually 10–seconds. There is always a trade-off between spatial resolution and acquisition speed. Modern systems use advanced algorithms to optimize this balance.Multiple display modes enhance interpretability. STIC volumes may be displayed in gray scale, color Doppler, HD flow and power Doppler, over which Volume Contrast Imaging (VCI) can be added. Adjusting slice thickness of VCI further improves visualization of intracardiac structures and flow patterns.

Figure 3.Modes of STIC acquisition and visualization

Modes of Visualization

One of the greatest strengths of 3D/4D fetal echocardiography is the variety of visualization tools available.

Multiplanar Mode: Multiplanar display simultaneously shows three orthogonal planes (A, B, and C). This allows reconstruction of standard views such as the four-chamber view, left ventricular outflow tract (LVOT), and right ventricular outflow tract (RVOT). It also enables coronal and sagittal reconstructions, precise measurements, and assessment of septal defects or valve pathology. For trainees and non-

experts, this mode significantly enhances spatial understanding.

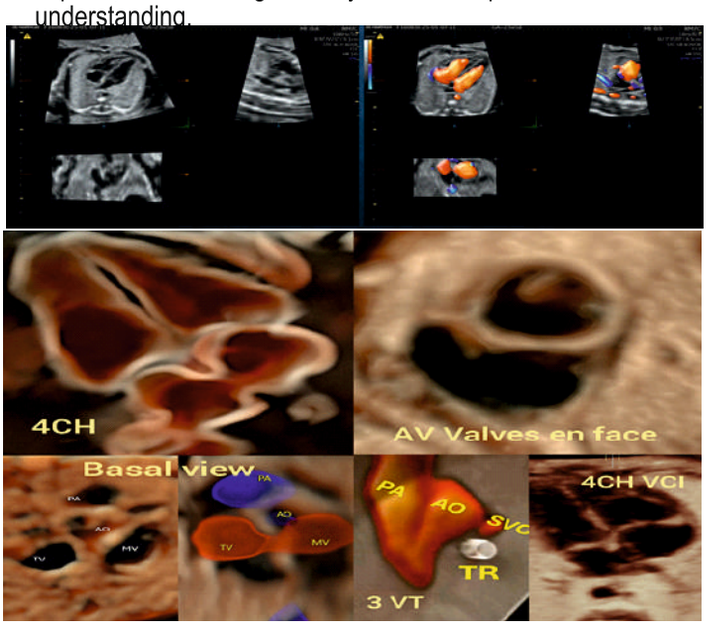

Figure 4, Initial multiplanar display of cardiac volume following acquisition and demonstration of rendered images of different cardiac views

Cine Loop (Temporal Rendering): The cine loop functionality plays the cardiac cycle as an animated sequence. This is particularly useful for evaluating valve motion and systolic–diastolic flow timing. It also serves as an excellent teaching and parental counseling tool because dynamic function is easier to appreciate than static images.

Tomographic Ultrasound Imaging (TUI): It displays sequential parallel slices similar to CT or MRI. This is extremely helpful in complex CHD, allowing systematic evaluation of vessel courses, anomalous pulmonary venous connections, and aortic arch abnormalities. Simultaneous comparison of multiple levels improves diagnostic confidence.

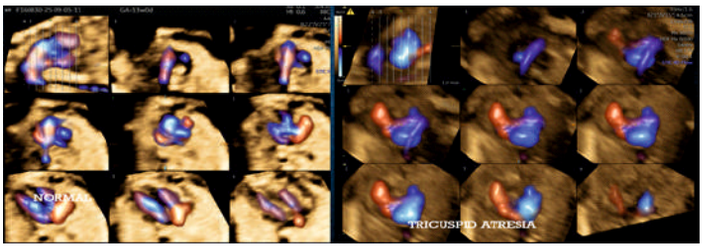

Figure 5. TUI mode showing a normal fetal heart (left) and a case of tricuspid atresia in a 13-weeks fetus(Right)

Inversion Mode: Inversion mode converts fluid-filled spaces into solid structures, thereby highlighting cardiac chambers and vascular lumens. This technique is particularly valuable in identifying aorta with neck branches, abnormal communications such as ventricular septal defects (VSD) and atrioventricular septal defects (AVSD).

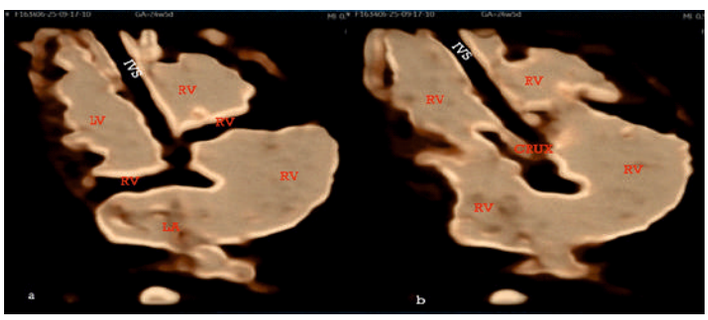

Figure 5 Normal 4 chamber view in inversion mode showing atrioventricular valves in systole (a) and diastole (b)

Rendering Mode: Surface and transparent rendering provide realistic 3D representations of cardiac morphology and blood flow relationships. These images are powerful for demonstrating spatial vessel relationships in complex lesions such as aberrant right subclavian artery (ARSA), transposition of the great arteries (TGA) or double outlet right ventricle (DORV). Rendering is also highly effective for parental counseling because it provides intuitive anatomical visualization.

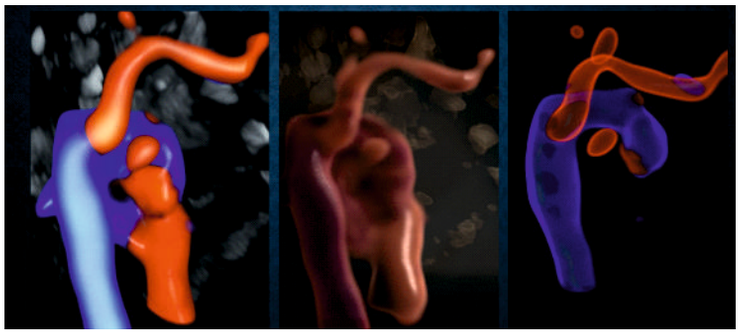

Figure 6. Typical origin of ARSA from anterior aspect of arch of aorta and its course shown in various color STIC rendering methods

Automation and Intelligent Navigation

A major recent advance is the introduction of intelligent navigation systems such as FINE (5D Heart) and Sono-VCAD. These tools automatically generate the nine standard fetal echocardiography planes. Automation reduces operator dependency, improves standardization, and allows less experienced sonologists to achieve consistent results. In regions with limited fetal cardiology expertise, this technology has significant potential impact.Electronic matrix probes further enhance performance. Probe containing thousands of independent elements that eliminate the need for mechanical sweep. Electronic beam steering enables rapid acquisition (approximately 1–3 seconds), improved temporal resolution, and real-time simultaneous orthogonal planes. Although currently expensive, this technology represents the future of real-time fetal cardiac imaging.

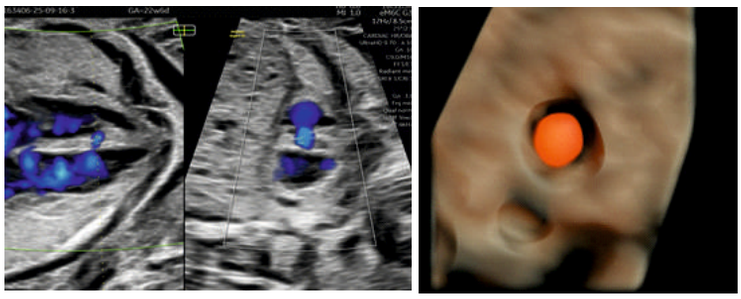

Figure 7 . Biplanar Display of a small muscular VSD by electronic matrix probe and 3D surface en-face view of VSD

Clinical Applications

The clinical utility of 3D/4D fetal echocardiography spans the entire gestational period.

First Trimester

Early cardiac assessment is increasingly feasible. STIC can identify major anomalies such as tricuspid atresia, hypoplastic left heart syndrome (HLHS), and pulmonary atresia with intact ventricular septum (PAIVS) even in the first trimester. Early detection facilitates timely counseling, genetic evaluation, and referral planning.

Second Trimester and Beyond

In the mid-trimester, 3D/4D imaging excels in characterizing structural heart disease. All major cardiac structural defects like ventricular septal defects , partial AVSD , AVSD , DORV ,

, transposition of the great arteries (TGA) , tetralogy of Fallot (TOF) and others can be detected confidently

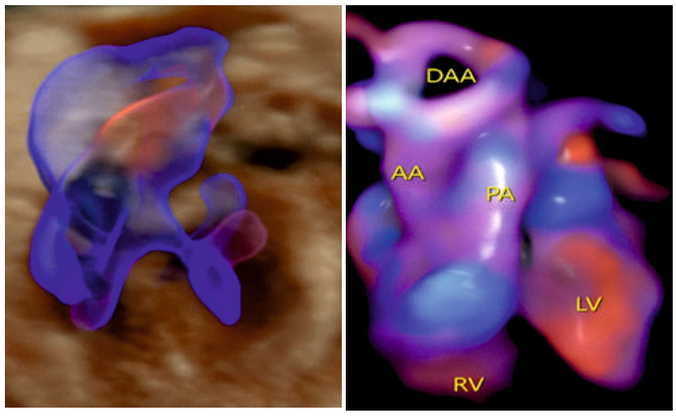

Figure 8 Demonstration of TOF (left) and doble aortic arch (right) by color STIC rendering

Outflow tract abnormalities and valve lesions are particularly well demonstrated. Examples include absent pulmonary valve syndrome , pulmonary stenosis, and Ebstein anomaly. The ability to appreciate spatial relationships between the great vessels is a major advantage over conventional 2D imaging.

Arch abnormalities are another area where 3D/4D imaging adds value. Clear depiction of interrupted aortic arch , aberrant right subclavian artery , double aortic arch , and right aortic arch is possible. These lesions often require multiplanar spatial understanding that benefits from volumetric imaging.Advanced applications include evaluation of the abnormal ductus venosus course, abnormal three-vessel view , aortopulmonary window , and coarctation of the aorta.

Pitfalls and Limitations

Despite its strengths, 3D/4D fetal echocardiography has limitations. The pitfalls slide motion artifacts from fetal or maternal movement, acoustic shadowing from ribs or spine, and reduced resolution in cases of maternal obesity. Proper patient positioning, probe adjustment, and optimized sweep angle remain essential.

Importantly, 3D/4D should be viewed as complementary to—not a replacement for—high-quality 2D fetal echocardiography. Acquisition of a good STIC volume still depends on obtaining an adequate 2D four-chamber view.

Future Directions

The future of fetal cardiac imaging is closely tied to artificial intelligence and advanced quantification. AI-assisted echocardiography may soon enable automatic detection and analysis of cardiac abnormalities. Quantitative tools such as fetal strain imaging, ejection fraction estimation, and myocardial performance assessment are likely to become more widely available.

Tele-echocardiography using stored STIC volumes is another promising development. Remote expert review could significantly improve diagnostic equity in resource-limited regions.

Practical Integration into Clinical Practice

For practicing obstetricians and fetal medicine specialists, the key question is how to integrate 3D/4D effectively. A pragmatic approach is to continue performing a standard 2D cardiac screening protocol, followed by STIC acquisition when the four-chamber view is satisfactory. Offline analysis can then be performed without prolonging patient scan time.

Training remains important. While automation is improving, understanding cardiac anatomy and spatial relationships is still essential for correct interpretation. Centers beginning their 3D/4D journey should focus first on mastering acquisition quality before exploring advanced rendering techniques.

Conclusion

Three-dimensional and four-dimensional ultrasound—driven by STIC technology—has significantly enhanced fetal cardiac assessment. The ability to acquire complete volumetric datasets, perform offline multiplanar analysis, and visualize complex spatial relationships makes it an invaluable adjunct to conventional fetal echocardiography.

Automation tools such as FINE/5D Heart are reducing operator dependency and expanding access to high-quality fetal cardiac screening. Meanwhile, emerging technologies including matrix probes, AI-assisted analysis, and tele-echocardiography promise further transformation.

However, the fundamental principle remains clear: 3D/4D ultrasound complements but does not replace meticulous 2D examination. When used judiciously and with proper technique, it markedly improves diagnostic confidence, teaching value, and parental counseling in fetal cardiology.

As fetal medicine continues to evolve, integration of advanced volumetric imaging into routine practice will likely become standard of care, ultimately improving prenatal detection and outcomes of congenital heart disease.

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Soft Markers, Hard Decisions: Interpreting EIF, SUA and cardiac risks

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SOCIETY OF FETAL MEDICINE

Second-trimester ultrasound often reveals subtle findings termed soft markers. These include echogenic intracardiac focus (EIF), single umbilical artery (SUA), choroid plexus cysts, echogenic bowel, urinary tract dilation, and mild ventriculomegaly. Soft markers are not structural anomalies but sonographic variants that may modify the probability of fetal aneuploidy. However, with the widespread use of first-trimester combined screening and cell-free DNA (cfDNA) testing, the interpretation of these markers has evolved significantly.

Changing Paradigms in Risk Stratification-

Historically, second-trimester soft markers were used to refine the risk of trisomy 21 and other chromosomal abnormalities. Current practice, however, emphasizes on the role of integrated risk assessment rather than isolated ultrasound findings. Current recommendations from the Society for Maternal-Fetal Medicine (SMFM) suggest that diagnostic testing should not be recommended solely on the basis of an isolated soft marker when prior serum or cfDNA screening is low risk.

The Fetal Medicine Foundation (FMF) approach similarly places greater reliance on first-trimester risk assessment using maternal age, nuchal translucency, nasal bone assessment, and biochemical markers. Within this framework, second-trimester soft markers function primarily as contextual modifiers rather than independent screening tools.

Echogenic Intracardiac Focus:

A Common Variant-

Echogenic intracardiac focus is one of the most frequently encountered soft markers at the mid-trimester anomaly scan. Although historically associated with trisomy 21, evidence indicates that isolated EIF confers only a minimal increase in risk, particularly when earlier screening is reassuring.

From a cardiac perspective, the ISUOG guidelines for fetal cardiac screening emphasize that isolated EIF does not significantly increase the risk of congenital heart disease. Consequently, additional fetal echocardiography is generally unnecessary unless other structural or functional cardiac abnormalities are identified.

Single Umbilical Artery:

The Importance of Isolation-

Single umbilical artery is another common finding at the anomaly scan. SUA may occur as an isolated variant or in association with structural anomalies, particularly cardiac or renal defects. The key clinical question is whether the SUA is truly isolated.

When SUA is detected, a detailed anatomical survey is essential to exclude associated abnormalities. Current guidance suggests that isolated SUA does not substantially increase the risk of aneuploidy but may justify fetal ECHO and third-trimester surveillance for fetal growth, given reported associations with fetal growth restriction and adverse perinatal outcomes.

Avoiding the Pitfall of False Positives-

Soft markers are relatively common in normal pregnancies. For example, isolated EIF may be observed in a small but notable proportion of structurally normal fetuses. In fact, it is a normal phenotypic finding in 8-10% of Asian population. Interpreting these findings without considering prior screening results can lead to unnecessary anxiety and potentially avoidable invasive testing.

With the increasing use of cfDNA screening, the emphasis has shifted toward contextual interpretation, where ultrasound findings are integrated with prior risk assessment and the overall fetal anatomical evaluation.

Counseling Without Creating Anxiety-

Perhaps the most challenging aspect of soft marker detection is communication with expectant parents. The terminology itself can provoke disproportionate concern. Effective counseling should focus on three key points:

- Most isolated soft markers represent normal variants.
- Risk assessment must incorporate prior screening results.
- Additional testing is rarely required

when screening is reassuring and the marker is isolated.

Conclusions-

Advances in prenatal screening have reshaped the clinical role of soft markers. Rather than serving as primary screening tools, they now function as contextual findings within a broader risk-assessment framework. For fetal medicine specialists, the challenge lies in integrating ultrasound findings with modern screening strategies while ensuring clear, balanced, and reassuring communication with families.

Soft markers may be subtle, but the decisions they prompt remain complex—requiring both scientific evidence and thoughtful counseling.

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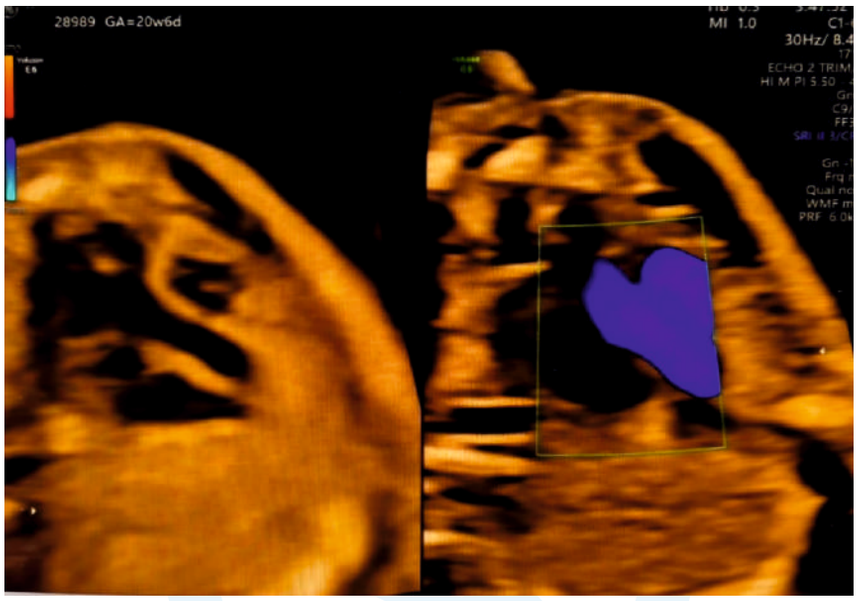

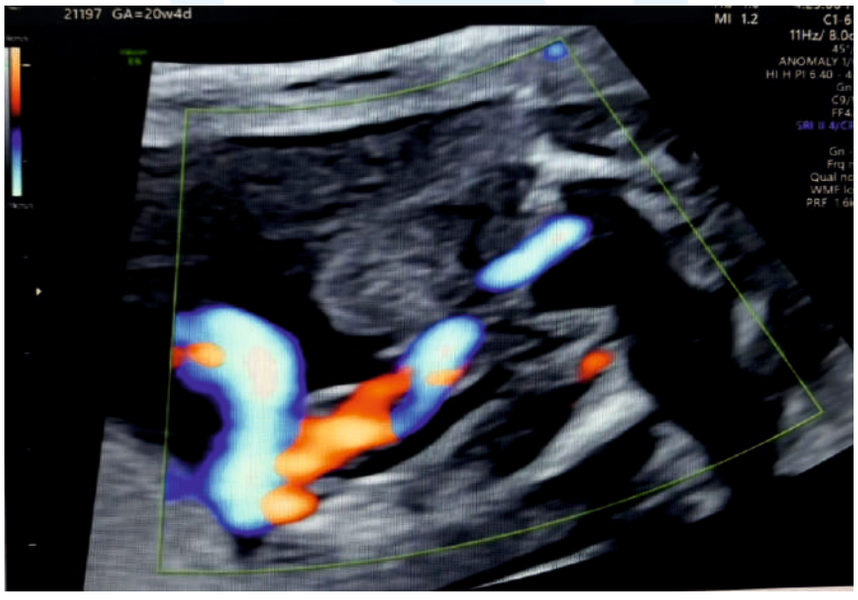

Dive Deeper

SOCIETY OF FETAL MEDICINE

Fetal Echo Indications: Catch the Clues

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Congenital heart disease affects approximately 8–10 per 1,000 live births and accounts for significant neonatal morbidity and mortality. While routine obstetric ultrasonography includes basic cardiac screening views, these do not detect all structural or functional abnormalities. Fetal echocardiography is an advanced, targeted ultrasound examination performed by trained specialists, typically between 18 and 22 weeks of gestation.

1. Maternal Indications

• Pre-gestational Diabetes: Mothers with Type 1 or Type 2 diabetes before pregnancy.

• Autoimmune Diseases: Mothers who test positive for specific antibodies (Anti-Ro/SSA or Anti-La/SSB), often associated with lupus or Sjögren's syndrome.

• Medication/Teratogen Exposure: Use of certain drugs like lithium, anti-seizure medications, ACE inhibitors, or retinoids during pregnancy.

• Infections: Exposure to infections such as Rubella (German measles), CMV, or Coxsackie during pregnancy.

• In Vitro Fertilization (IVF): Use of assisted reproductive technology is often considered an indication.

2. Fetal Indications

• Abnormal Obstetric Ultrasound: Suspicion of a heart defect or an abnormal heart rhythm (arrhythmia) during a routine scan is the most common reason for referral.

• Increased Nuchal Translucency: A thickened nuchal fold (typically ≥ 3.5 mm) seen in the first trimester.

• Extracardiac Anomalies: Major abnormalities in other organs, such as the kidneys, brain, or bones.

• Chromosomal Abnormalities: Genetic conditions like Down syndrome (Trisomy 21) or other karyotype abnormalities.

• Fetal Hydrops: Abnormal fluid accumulation in two or more fetal compartments.

• Twin Pregnancies: Specifically monochorionic (identical) twins, due to the risk of twin-to-twin transfusion syndrome (TTTS).

3. Familial and Genetic Indications

• A previous child with congenital heart disease

• Parental history of CHD

• Known genetic syndromes with cardiac associations (e.g., 22q11.2 deletion syndrome, Noonan syndrome) warrant referral for fetal echocardiography.

4. Technical and Screening-Based Indications

Suboptimal cardiac visualization, abnormal Doppler findings, or suspected cardiac tumors are additional indications for specialized cardiac evaluation.

Clinical Impact

Early prenatal diagnosis enables multidisciplinary counselling, delivery planning in tertiary centres, and immediate neonatal cardiac intervention, significantly improving survival in critical CHD cases.

Conclusion

Fetal echocardiography is essential for high-risk pregnancies and suspected fetal cardiac abnormalities. Appropriate identification of indications enhances early detection and improves perinatal outcomes.

Dive Deeper

SOCIETY OF FETAL MEDICINE

SOCIETY OF FETAL MEDICINE

The Art and Physics of the Fetal Heart: Optimizing Image Quality in Echocardiography

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In fetal medicine, the heart remains the most elusive and technically demanding organ to image. It is a small, rapidly moving structure, shielded by fetal ribs and subject to the vagaries of maternal habitus, fetal position, and liquor volume.

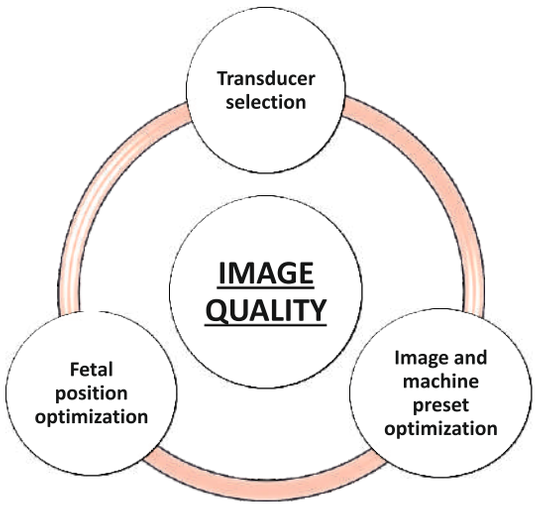
The difference between a missed diagnosis and a life-saving intervention often comes down to a few millimeters of probe movement or a decibel of gain.

While clinical acumen is vital, a specialist's ability to "interrogate" the heart depends heavily on mastering the ultrasound machine's physics.

Every fetus requires a bespoke approach. The variables we can control—our machine settings and ergonomics—will be underutilized, if we rely solely on the factory presets.

This article aims to bridge the gap between basic scanning and the high-level precision of a fetal echocardiogram.

Let us elaborate the "physics-to-fingertip" connection.



1. The Foundation: Ergonomics and Probe Positioning and Selection

Before touching any knob, we must address the physical interface. Image quality begins with the transducer's relationship to the fetal anatomy.

a) The "Perpendicularity" Rule: Ultrasound waves reflect best when they hit a surface at a 90-degree angle, that is your beam must be perpendicular to the structure you want to see. Even little deviation can cause significant "drop-out" artifacts.

b) The Acoustic Window: Always hunt for the thinnest part of the maternal abdominal wall. Often, sliding the probe laterally to find a "thinner" window is more effective than increasing the gain.

c) Pressure Management: Consistent, firm yet gentle pressure displaces intervening bowel gas and reduces the distance the beam must travel, thereby reducing attenuation.

d) Frequency Selection (The Resolution vs. Penetration Trade-off):

Optimal scanning starts with the transducer. All transducers have different operating frequencies and capabilities.

High frequency transducer provides superior detail resolution but, of course, with limited sound penetration. These frequencies can be applied in all trimesters, however best used for early anatomy scans (12–16 weeks) or thin patients.

For patients with a high body mass, scanning in the late second trimester/third trimester, in cases of polyhydramnios syndrome, or even when there is rib shadowing, you may be forced to drop the frequency to actually "reach" the heart, sacrificing some detail, with a low frequency transducer.

Hence, select a transducer wisely that allows for adequate penetration and optimal resolution.

2. The Temporal Resolution Challenge: Maximizing Frame Rate

Temporal resolution is the ability to locate moving structures at any particular instant in time.

The fetal heart beats between 120 and 160 times per minute. To capture the rapid movement of the valve leaflets or the subtle motion of the interventricular septum, high temporal resolution (expressed as Frame Rate, HZ) is non-negotiable.

If the frame rate is too low, the image will appear "laggy," and small defects like a peri membranous VSD may be missed.

Better the temporal resolution, improved is the detailed resolution.

To maximize temporal resolution, the machine must process fewer data points per frame. Here is how the image presets need to be optimized so as to maintain a high frame rate of greater than 25 frames per second.

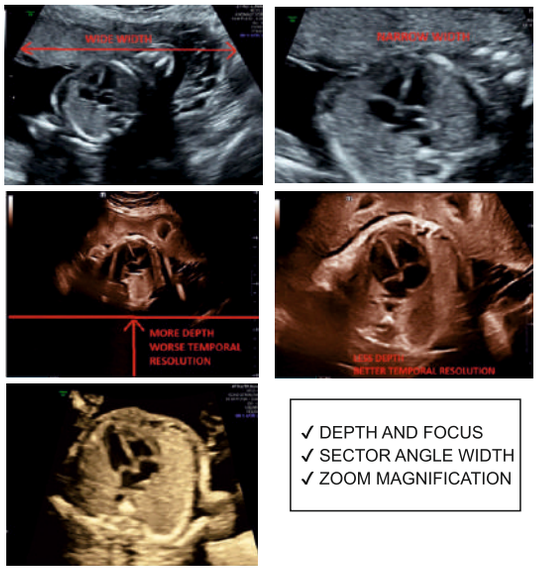
a) Narrow the Sector Width: This is the most effective way to increase frame rate. Instead of scanning a wide 90° field, narrow the field to just the width of the heart.

A narrow sector width decreases the number of lines required to produce the image; this decreases the frame time thereby increasing the frame rate and improving temporal resolution.
The perceived image resolution is also improved by removing extraneous information from the image, allowing your eyes to focus on the fetal heart.

b) Reduce Depth: Do not leave "dead space" behind the spine. Set the depth so that the heart fills the screen. By decreasing the depth, the sound beam can get to the max depth quicker. This decreases the line time, which decreases the time it takes to produce each frame. The result is an increase in the frame rate leading to improved temporal resolution.
Visualization of the heart is also better by making it larger and improving the perceived resolution.

c) Magnification (Read vs. Write Zoom): There are two different methods to achieve magnification of the image. Each method has different effects on image resolution.
Read Zoom is performed by simply increasing the size of the entire image. Read Zoom only enlarges the existing pixel; hence the original image resolution remains unchanged.
For Write Zoom the area to be magnified is selected and then the ultrasound system will rescan the new smaller area but with the line density utilized for a whole image. The increased line density over the smaller area results in improved detail resolution.
Always use Write Zoom (High-Definition Zoom).

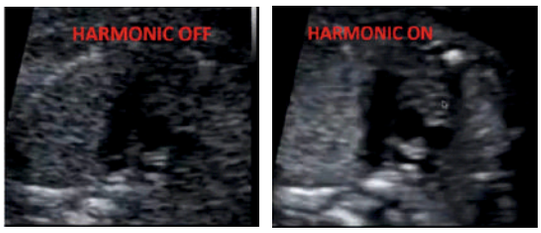
d) Minimize Focal Zones: While multiple focal zones improve detail across the whole screen, they effect the temporal resolution. When looking at the 4-chamber heart, a single focal zone is desirable.



3. Spatial Resolution: Achieving the "Crisp" Cardiac Border

Where temporal resolution is about speed, spatial resolution is about clarity and detail.

Spatial resolution is the ability to distinguish two separate points as distinct. In the heart, this means seeing the thin insertion of the tricuspid valve or the integrity of the atrial septum.
a) Harmonic Imaging (THI): The theory behind harmonic imaging is that the beam is transmitted out at a low frequency and the return beam is received at twice the transmitted frequency.
This acts to improve detail resolution but maintain adequate penetration. The blood-filled ventricles appear truly anechoic (black) and the endocardial borders sharper.



b) Dynamic Range (Compression): High contrast equates to a low compression setting (low dynamic range) which means fewer shades of grey will be represented. Conversely, low contrast utilizes many shades of grey thus requiring a high compression setting (high dynamic range).
The fetal heart has a relatively high contrast. Hence, a lower dynamic range (higher contrast) is generally preferred for the heart. This emphasizes the interface between the blood and the myocardium, making valve movement more apparent.

4. Optimization for Color and Spectral Doppler

Doppler is the cornerstone of functional cardiac assessment. However, it is also the most common source of "noise" and diagnostic error.

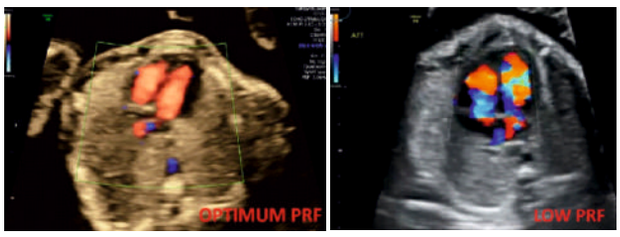
The Color Doppler Setup

a) The Box Size: Just like the sector width, the color box should be as small as possible. A large color box requires massive processing power, which severely slows down the frame rate and creates "bleeding" artifacts. Hence, make the color box as small as possible, covering only the area of interest.

b) Color Gain: Color gain should initially be set on low (ie, less color) and gradually increased until amount of color is optimized.

c) Scale/PRF (Pulse Repetition Frequency): Defined as the number of ultrasound pulses emitted by the transducer per second.
In the context of fetal echocardiography, optimizing PRF is essential to distinguish fast-moving fetal blood flow from low-velocity movements without causing aliasing or noise.
If the color scale is too low for high-velocity flow, aliasing occurs. If the scale is too high, low-velocity flows (like the pulmonary veins) will not be detected.

High-velocity flow (>50 cm/sec)	Low-velocity flow (<30 cm/sec)
Atrioventricular valves	Pulmonary veins
Semilunar valves	Bicaval (IVC/SVC)
The great vessels (3VV)	Evaluating atrial and ventricular septum



The Spectral Doppler Setup

a) Angle of Insolation: This is the "golden rule" of fetal echocardiography. For accurate velocity measurements (e.g., across the aortic valve or in the ductus arteriosus), the beam must be as close to 0° (parallel to flow) as possible.

b) Sample Volume (Gate): For valvular assessment, use a gate of 2–3 mm. A larger gate will pick up "contamination" from adjacent vessels, making the waveform difficult to trace.

5. Specialized Machine Tweaks: The "Secret" Dials

Beyond the standard knobs, these features can drastically improve the diagnostic yield.

a) Speckle Reduction Imaging (SRI): Eliminates speckle – weak signals, and strong signals are intensified and brightened, leading to smoother cleaner image. Use a moderate setting. Too much SRI can "blur" the heart and make it look like a cartoon, potentially hiding a small VSD.

b) Persistence: High persistence averages frames over time, which "smooths" the image but creates a ghosting effect that obscures the rapid movement of the heart valves. In echocardiography, persistence is generally turned low.

c) Scanning in different tones: Human eye is able to differentiate details better in color rather than gray scale. While the classic “black and white” is the gold standard, switching tones to sepia/blue/cyan tones may actually help eyes pick up details that might be lost in a standard grayscale.

6. The "Acoustic Challenge": Optimizing Fetal Echo in the High-BMI Patient

Maternal obesity represents a significant technical barrier. Adipose tissue acts as a powerful acoustic filter, causing absorption and refraction of the beam. Increased distance between the transducer and fetal anatomy causes degraded resolution.

To achieve a diagnostic fetal echocardiogram in these cases, we must move beyond standard presets and apply specific "rescue" settings to maximize penetration without sacrificing the critical temporal resolution needed for cardiac work.

a) Frequency Modulation: Downshift to low frequency (3–5 Mhz). Lower frequencies have longer wavelengths that navigate adipose tissue with less energy loss.

b) Fundamental Mode: While THI is the gold standard for thin patients, it can be counterproductive in the very obese. If the image is too "soupy," switch back to Fundamental Mode to regain signal-to-noise ratio.

c) The "Window" Hunt: Don't be afraid to scan from the flanks. Adipose layers are often thinner laterally than at the midline. Consistent, firm pressure can also displace intervening tissue and reduce the distance the beam must travel. Scanning above/below the panniculus also helps.

7. The Digital Revolution: The Role of Artificial Intelligence (AI)

As we move into 2026, the "AI-enhanced" echo is no longer a futuristic concept but a clinical reality. AI is transforming fetal echocardiography from an operator-dependent art into a standardized science.

a) Automated View Acquisition: Recent FDA-cleared tools (like BrightHeart's B-Right Views) now assist in the real-time detection of the five standard cardiac views. These systems provide immediate feedback, alerting the specialist if a view is sub-optimal or if specific structural markers are missing.

b) Intelligent Navigation: Technologies like FINE (Fetal Intelligent Navigation Echocardiography) allow the machine to automatically extract standard views from a 3D volume, reducing the time spent manual-searching for the "perfect" plane.

c) Quality Control & Measurement: AI algorithms now automate routine biometry—measuring heart-to-chest ratios and chamber dimensions with a reproducibility that exceeds human capability. This reduces fatigue and allows the specialist to focus on complex diagnostic decision-making rather than repetitive measurements.

d) Closing the Diagnostic Gap: AI is particularly transformative in high-BMI scanning and low-resource settings, where it helps by identifying subtle cues of congenital heart disease (CHD) that might be masked by acoustic noise.

Parameter	Recommended se□ng for heart	Why?
Sector width	Narrow (30-45 degree)	Maximizes frame rate
Focal Zone	Single, at valve level	Prevents temporal lag
Harmonics	On	Clears noise from chambers
Dynamic range	Low (45 – 55 dB)	Enhances contrast of endocardium
Persistence	Low or off	Prevents ghosting of fast moving valves
Color box	Mimimal size	Maintains high frame rate

In nutshell,

If you see	The probable cause is	The Clinical fix
"Laggy" or jerky motion of the heart valves	Low Frame Rate: The machine isprocessing too much data per second.	Narrow the sector width (reduce the "pie slice") and use a single focal zone.
Ghosting" or trails behind moving structures (e.g., leaflets).	High Persistence: The machine is averaging too many previous frames.	Turn Persistence to 'Low' or 'Off' to capture true real time motion.
Chamber "Clutter" (haze inside the ventricles).	Side Lobe Artifacts: Secondary ultrasound beams are creating noise.	Increase Harmonic Imaging and lower the nearfield TG(sliders.
Color "Bleeding" outside the vessel or valve walls	Excessive Gain: The color sensitivity is set too high for the flow.	Reduce Color Gain or Increase the PRF (Scale) to match the velocity.
Poor Velocity Signal or weak Doppler waveforms.	High Insonation Angle:The beam is hitting the blood flow at a steep angle.	Re-position the probe to ensure the beam is as parallel to flow as possible close to 0°).

Ultimately, optimizing a fetal echocardiography is a dynamic process that extends far beyond standard presets. A great fetal heart scan isn't just about having an expensive machine; it’s about knowing which buttons to turn, and ability to master the machine’s specific functions.

By narrowing focus, adjusting the frequency and leveraging the entire maternal abdomen for the best acoustic window, we can see through the "noise" and get the clear answers our patients need.

In the end, these small tweaks lead to better diagnoses and better care for the families we serve!

SOCIETY OF FETAL MEDICINE

RV: LV Discrepancy - Overdiagnosis of COA vs Vigilance

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The coarctation of aorta (CoA) is characterized by narrowing of the thoracic aorta at the insertion of the arterial duct, with or without additional abnormalities of the aortic arch.1 CoA accounts for approximately 8% of congenital heart diseases (CHDs) and occurs in 0.2 to 0.62 per 1000 live births; and is a cause of remarkable morbidity and mortality if not diagnosed early.1,2 Upon birth and after ductal closure, a critical CoA will cause an increase in left ventricular afterload with severe cardiac failure, poor perfusion of the lower part of the body, end-organ damage and death.3 Therefore, the diagnosis of prenatal CoA improves perioperative outcome by allowing planned delivery in a tertiary care center and early institution of prostaglandin treatment to prevent the closure of the duct.4,5 However, in-utero diagnosis of CoA still remains challenging and is affected by a high rate of false positives, especially during the third trimester, because it relies on indirect and non-specific signs, such as cardiac asymmetry with right dominance that may also be seen in normal fetuses in late pregnancy.6 The overall detection rate of prenatal ultrasound in identifying this anomaly has been reported to be poor at the time of the routine anomaly scan. Moreover, routine third-trimester scan is not universally performed, unless fetal or maternal complications are suspected, and it is usually performed almost exclusively to assess fetal growth. In this scenario, the presence of cardiovascular disproportion may be easily overlooked, thus explaining the reported low detection rate for CoA. The definition of cardiovascular disproportion is usually subjective, and structurally normal fetal hearts in the third trimester of pregnancies exhibit a slight degree of physiological disproportion. Conversely, disproportion detected in the late second or early third trimester of pregnancies carries an increased risk for the occurrence of CoA. False-positive diagnosis can result in parental anxiety and unnecessary stress. Therefore, it is necessary to have specialized ultrasonographic tools for the correct diagnosis of coarctation. The differential diagnosis of isolated 4-chamber cardiac disproportion is also wide. Due to the pre- and post-natal screening system limits, many institutions adopted a cautious postnatal management plan once antenatal suspicion is raised, although contributing to the increase in false positive cases. This is warranted considering the severity of the disease if left undiagnosed, but the unnecessary admission of the neonate with the suspicion of CoA to the cardiac or neonatal intensive care unit may contribute to family distress due to the increased medicalization of an otherwise normal infant. The use of cardiovascular disproportion alone has an overall low diagnostic accuracy that is even lower during the third trimester. Several ultrasound signs have been proposed to potentially improve the detection rate of prenatal diagnosis for CoA.

LV/RV discrepancy in evolving COA (Figure 1):

-Method:

1 .Left ventricular and right ventricular (LV/RV) diameter ratio: Transverse diameters of the RV and LV under the level of atrioventricular valves, at the end of the diastole in 4 chamber view are measured.

2. LV/RV length ratio:

- Diagnostic challenge: Fetal COA causes reduced left sided flow, leading to smaller LV and a dominant, larger RV. However, because the ductus arteriosus diverts most fetal blood away from the isthmus, many cases do not present with significant obstruction until after birth.

-Sensitivity and timing: Discrepant ventricular size has only a moderate sensitivity and a low specificity and low positive predictive value for the diagnosis.7 Ventricular discrepancy is most sensitive in the second trimester (before 25 weeks). After 34 weeks, a false positive rate as high as 80% is reported.

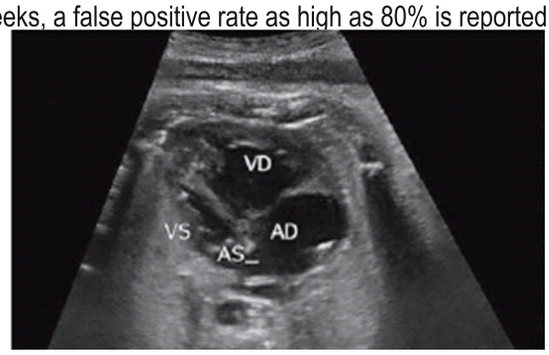

Figure 1: 4-chamber view showing ventricular disproportion. AS= Left atrium, AD- right atrium, VS=Left ventricle, VD= Right ventricle.

[Image collected from Mărginean C, Mărginean CO, Muntean I, Togănel R, Voidăzan S, Gozar L. The role of ventricular disproportion, aortic, and ductal isthmus ultrasound measurements for the diagnosis of fetal aortic coarctation, in the third trimester of pregnancy. Med Ultrason. 2015 Dec;17(4):475-81.]

Other key indicators (Beyond discrepancy):

Cardiac dimensions are measured at their maximum size and measurements are taken from the inner edge to the inner edge. All z scores are computed on gestational age (GA) and femur length.

1. Mid-cavity ratio: a LV mid-cavitary dimension to RV mid-cavitary dimension (LVmc/RVmc) of <0.6 provides a 70% sensitivity and 67% specificity for predicting postnatal intervention.9
2. MV and TV Z-score and its ratio: The diameter of the atrioventricular valves are measured in apical four-chamber view.
3. Aortic valve (AoV) Z-score and Pulmonary valve (PV)/AoV diameter ratio: PV/AoV ratio >1.6 is significantly associated with CoA and it has good sensitivity (86.2%), but a low specificity (51.8%).10
4. Main pulmonary artery (MPA) and ascending aorta (AAO) diameters, Z-scores and ratio:
A COA is almost always associated with a discrepancy of the great vessels where the diameter of the arteria pulmonalis is bigger than the diameter of the aorta during diastole. This is probably related to blood flow redistribution because of the increased resistance of the LV outflow tract. it can be only a temporary redistribution that normalises after birth, or a permanent redistribution seen in cases requiring cardiac surgery after delivery. Ascending aorta (Ao), pulmonary artery (PA) in 3 vessel section. For a PA:Ao ratio of 1.60 or more, the sensitivity was 83%, specificity 85%, positive predictive value 62.5% and negative predictive value 94%.11
5. Distal transverse aortic arch (TAA) and Aortic isthmus (AoI) diameter and Z-score: The internal diameter of the AoI is measured immediately proximal to the insertion of the arterial duct in the three vessels and trachea (3VT) view (Figure 2). The distal TAA diameter is also acquired in the 3VT view. AoI Z score ≤ -2 in 3 vessel view (3VV) or sagittal view by fetal echocardiography requires surveillance in prenatal life and surgery in postnatal life

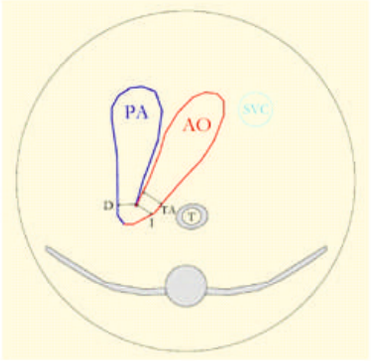

Figure 2: The diagram reproduces the three vessel and the tracheal view and shows the pulmonary artery (PA) leading into the arterial duct (D), the aortic arch (AO), and the superior vena cava (SVC). The lines show the measurement of, respectively, the isthmus (I), the duct, and the distal transverse aortic arch (TA) diameter.

Diagnosis of an aortic isthmus coarctation is possible in 45% of the cases during the fetal life using gray scale ultrasonography12 and the aortic arch < 3 mm could be considered a good predictor for the need of neonatal surgical intervention (Figure 3).

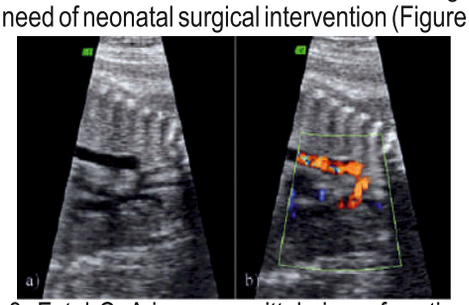

Figure 3: Fetal CoA in parasagittal view of aortic arch and the descending aorta: a) gray scale, b) color Doppler.
[Image collected from Mărginean C, Mărginean CO, Muntean I, Togănel R, Voidăzan S, Gozar L. The role of ventricular disproportion, aortic, and ductal isthmus ultrasound measurements for the diagnosis of fetal aortic coarctation, in the third trimester of pregnancy. Med Ultrason. 2015 Dec;17(4):475-81.]

The measurement of the aortic isthmus during fetal ultrasound is one of the most important criteria for the diagnosis of CoA. Therefore, Achiron et al.14 describes normal values of more than 3 mm of this lumen after 28 weeks, and more than 4.2 mm after 32 weeks, and concluded that lower values than the above can raise the suspicion of CoA.
6. Isthmus/ ductal ratio:
Ao isthmus (AoI) and arterial duct (AD) are measured near to their confluence in 3 vessel view. This ratio <0.74 is highly suspicious for COA.15 Serial isthmus-to-ductal ratios can also help distinguish fetuses that would require surgery from those requiring surveillance. The higher the scores are (thus the higher the ratio), the less likely a CoA exists (Figure 4)

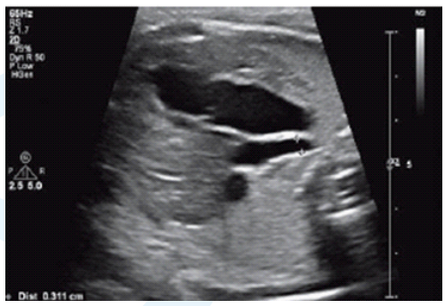

Figure 4: 3VT View with the aortic arch smaller than the ductus arteriosus and suspicion of fetal CoA.
[Image collected from Mărginean C, Mărginean CO, Muntean I, Togănel R, Voidăzan S, Gozar L. The role of ventricular disproportion, aortic, and ductal isthmus ultrasound measurements for the diagnosis of fetal aortic coarctation, in the third trimester of pregnancy. Med Ultrason. 2015 Dec;17(4):475-81.]

7. Aortic/ ductal angle: In normal fetuses the aortic/ductal angle ranges from 128.2° to 167 degrees. Smaller angle (82.2-125º) is characteristic of COA.16

8. Reversed or mixed flow at aortic arch: Retrograde flow in the transverse arch during systole is a key marker. Retrograde flow in the aortic arch is not always present and can be physiological in third trimester. Retrograde flow in patients with coarctation is mainly observed during systole. Therefore, retrograde flow in the fetal aortic arch combined with a small left heart is suspicious for coarctation.17

9. Bidirectional flow through foramen ovale: Foramen ovale membrane was defined as redundant when it herniated into the left atrium for more than 50% of the left atrial diameter. Bidirectional flow through FO on color Doppler can diagnose 65% of CoA cases.18,19 This was not identified as strong prenatal CoA predictors in the multivariate logistic analysis. In fact, these functional parameters may also be detected in the presence of an isolated redundant foramen ovale membrane that may mimic fetal aortic coarctation, especially during the third trimester of pregnancy, increasing the rate of false positive CoA diagnosis.

Pasquini et al. used Z-score of AoI and AD diameter in the prenatal prediction for CoA especially after 26 weeks of pregnancy.15 According to Mărginean et al.20 a combination of RV/LV<1.5, aortic isthmus (AoI) <4.2 mm, and arterial duct (AD)/AoI >1.4 gave the overall best predictive accuracy for CoA. Gomez et al considered that the ventricular and great arteries disproportion were indirect signs of CoA. They composed a multiparametric score for CoA prognosis, the best combination for CoA prediction being the pregnancy age of diagnosis, Z score of the ascending aorta, ratio between the size of the pulmonary and aortic valves, respectively the Z-score of the aortic isthmus, measured in the three vessels section. Using this score the diagnosis accuracy was enhanced from 20% to 51% after the age of 28 weeks.21 Another study showed the AV and distal TAA z-scores respectively had strongest association with postnatal CoA.22 These parameters were shown to have the best balance between sensitivity and specificity for the diagnosis of CoA in fetuses with isolated cardiac asymmetry. This study has stratified the risk of developing CoA after birth into low, moderate, or high based on obstetrical history (i.e., family history of CoA, early suspicion of cardiac asymmetry during the ultrasound scan in the second trimester) and major echocardiographic parameters detected at the last fetal echocardiographic examination. Low CoA risk fetuses received the first diagnosis of CoA suspicion during the third trimester of pregnancy (> 28 weeks of GA, late diagnosis), had an aortic isthmus narrowing (-3< isthmus z-score ≤-2; 0.5< AI/AD ratio <0.7) but a good size transverse aortic arch at the subjective analysis, and a mild discrepancy between the right and left side structures (LV/RV ratio ≥0.6). Moderate CoA risk fetuses received an early diagnosis (<28 weeks of GA), had an aortic isthmus narrowing (isthmus z-score ≤-3; AI/AD ratio ≤0.5), and a more evident discrepancy between left and right side dimensions both at the level of the ventricles (mitral z-score <-3, LV/RV ratio <0.6) and of the great arteries (MPA/AA ratio 2) compared to the low CoA risk fetuses. High risk fetuses presented moderate-risk anatomic features with marked hypoplasia of the aortic arch (distal TAA z-score <-1), a borderline LV with MV hypoplasia (z-score <-5), and an aortic arch flow reversal and bidirectional flow at the level of the foramen ovale.

Other associated factors:

1. Bicuspid aortic valve- almost never detected prenatally, sometimes it can be suspected by a poststenotic dilatation or by abnormal Doppler flow.
2. Aortic valve stenosis
3. Large VSD
4. Mitral stenosis
5. Persistent left superior vena cava (LSVC)
6. Chromosomal abnormalities, especially when associated with other anomalies (14% of patients with Turner syndrome associated with COA).

"Evolving" Coarctation and clinical vigilance:

"Evolving" implies that a previously normal-appearing arch may develop significant constriction postnatally as the ductus arteriosus closes.

Sudden collapse: Neonates with suspected COA (especially when large PDA, PFO/ASD and VSD are present) can experience sudden collapse within 12 hours of birth due to rapid ductal closure.

High vigilance: Current literature emphasizes that even when prenatal suspicion is low, the presence of LV/RV discrepancy necessitates high vigilance in the neonatal unit.

Alternative causes:

Other, often more severe lesions must be considered, including hypoplastic left heart syndrome (HLHS), aortic valve stenosis, or transposition of great arteries (TGA).

Recent findings and emerging techniques:

1. Novel diagnostic model: Recent studies propose combining morphology (e.g., aortic isthmus/VSD ratio) with functional variables like left ventricular longitudinal strain (LVLS), which has shown high accuracy (AUC 0.96) in predicting postnatal COA.
2. 3D/4D imaging: Coarctation shelf is visualized using 3D ultrasound and static 3D volumes of the fetal heart with colour flow mapping.23 Visualization of a narrowing of the isthmus and tortuosity of the aortic arch using bidirectional high-definition flow combined with spatiotemporal image correlation (STIC).17

- B-flow imaging: B-flow is a display modality in 4D sonography that enhances signals from weak blood reflectors from vessels, suppressing strong signals from surrounding tissues, and is angle independent.24
- Multiplanar imaging- 3D and 4D sonography using a combination of spatiotemporal image correlation (STIC) and tomographic ultrasound imaging (TUI) is a novel display modality that allows for the simultaneous visualization of 3 orthogonal anatomic planes. The use of the anatomical planes can help for the examination of the fetal heart with the possibility of off-line use and the reduction of operator dependency.23
- To measure the sizes of both pulmonary artery and aorta in the three-vessel view by using 4-D sonography with STIC. PA:Ao ratio is significantly higher in fetuses with CoA compared to those with a normal heart.25

Key takeways for clinical practice:

1. Vigilance is key: a "smaller" left heart is a red flag. A prenatal diagnosis of "suspected COA" warrants delivery at a center with pediatric cardiac intensive care.
2. Multimodal evaluation: do not rely on 4-chamber view alone. Evaluate 3VT view, isthmus/ductal ratio, and Doppler flow across the arch.
3. Third trimester trap: be aware that physiological RV dominance after 34 weeks can mimic COA, but any discrepancy warrants close observation, not immediate assumption of a false positive.

Detecting arch hypoplasia in fetal life is well feasible but it is also important to try to diagnose fetal coarctation because the prenatal diagnosis of this cardiac defect improves survival and reduces neonatal morbidity, at least if neonates are born in a center with specialised cardiac care.

Conclusion:

Antenatal detection of Coarctation of aorta still remains a challenging subject. Ventricular discrepancy is definitely the "red flag', if identified in 2nd trimester and should not be ignored. However, isolated ventricular discrepancy if taken as sole criteria for predicting Coarctation of aorta that can be misleading due to high false positivity rate. Which only increase unnecessary parental anxiety and misuse of resources.

Fetal cardiologist should try to identify hypoplasia of aortic arch along with ventricular discrepancy. But identification of arch hypoplasia in fetus is still not accurately defined. Till getting accurate full prove diagnostic criteria by multicentric study, fetal cardiologist should spend time and adopt describe criteria's available in literature.

Formulate own institutional guideline and follow up protocol for suspected evolving coarctation of aorta. Till date vigilance is better than losing even a single newborn due to missing otherwise correctable lesion.

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SOCIETY OF FETAL MEDICINE

Fetal Arrhythmias - SVT, Heart Blocks & Hydrops: How to Approach?

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Introduction: Fetal heart rate ranges between 110 and 160 beats per minute. It is considered abnormal if the heart rate goes beyond the normal ranges or the rhythm becomes irregular. Premature atrial contractions (PAC) are the commonest of all fetal arrhythmias but are hardly of any clinical significance. Clinically significant fetal arrhythmias occur in about 1 per 4000 live births (1). This constitutes 10% of all fetal arrhythmias and requires continued surveillance and monitoring. While supraventricular tachycardia (SVT) is the most commonly encountered fetal arrhythmia, others such as atrial flutter, ventricular tachycardia, and bradyarrhythmias such sinus bradycardia and heart blocks are not uncommon and may indicate severe underlying disorders (1). It complicates around 1–3% of pregnancies and accounts for 20% of referrals to paediatric cardiologists (2).

Clinical presentation:

Fetal arrythmias are commonly detected in uncomplicated pregnancies during a routine obstetric scan. They may be intermittent or sustained and seldom result in fetal hemodynamic compromise resulting in low cardiac output, fetal hydrops, and eventually fetal demise. Fetuses with hydrops must be re-evaluated and re-assessed at regular intervals to rule out intermittent fetal arrhythmias.
Connective tissue disorders in the mother such as Systemic Lupus Erythematosus or Sjogren's syndrome are commonly associated with complete heart block (CHB) in the fetus. There may also be an underlying cardiomyopathy or structural heart disease like cardiac tumours which should be evaluated thoroughly in every case of fetal arrhythmia. The combination of a sustained arrhythmia, structural heart disease, and hydrops fetalis carries a very poor prognosis.

Prenatal diagnosis of arrhythmias:

Fetal electrocardiography (fECG) and fetal magnetocardiography (fMCG) are principal tools for diagnosing fetal arrhythmias. fMCG is a noninvasive method that records the fetal heart's electrical activity in a manner similar to a conventional electrocardiogram (ECG). It enhances the evaluation of fetal cardiac electrophysiology by enabling detailed analysis of T-wave morphology and accurate measurement of the QT interval. This modality is particularly useful for clarifying the mechanisms of tachyarrhythmias and for detecting high-risk conditions, such as long QT syndrome. Nevertheless, fMCG is costly and, in our country, is mainly confined to academic settings. Given its limited clinical accessibility and practicality, fetal cardiac rhythm is more commonly assessed using Doppler ultrasound (3).
During routine obstetric evaluation, Doppler assessment of pulsatile fetal cardiac flow can determine the ventricular rate, but it does not reliably define the atrioventricular (AV) relationship or identify the site of rhythm origin (Figure 1). A more appropriate technique is M-mode (motion mode) echocardiography. The M-mode cursor should be positioned to simultaneously transect the atrial and ventricular walls, allowing visualization of both atrial (A) and ventricular (V) contractions and assessment of their temporal relationship (Figure 2) (4). For instance, Doppler interrogation across these walls may reveal a 2:1 AV block, where two atrial contractions correspond to one ventricular contraction. Alternatively, the M-mode cursor may be aligned between either atrium (right or left) and a semilunar valve (aortic or pulmonary). This method also allows evaluation of the AV relationship and facilitates measurement of the AV and VA intervals, fetal heart rate, and AV conduction patterns. However, M-mode evaluation has limitations. It is often less reliable in the first trimester and in challenging circumstances such as unfavorable fetal positioning or in hydropic fetuses, where obtaining clear recordings of atrial and ventricular wall motion can be difficult. A systematic approach is essential to obtain the correct diagnosis in patients presenting with symptoms compatible with an arrhythmia. The differential diagnosis of atrial arrhythmias is given in table 1 (adapted from (1).

Atrial and Ventricular Ectopy:

Premature atrial contractions (PACs) are the most common fetal arrhythmia and may appear as isolated beats or in patterns (bigeminy/trigeminy). They usually require no treatment, but because 1–2% progress to supraventricular arrhythmias, close monitoring is recommended.
Premature ventricular contractions (PVCs) may also occur singly or in patterns but warrant evaluation for cardiomyopathy, cardiac tumours, structural defects, and inherited arrhythmias, with close prenatal and postnatal follow-up.

Fetal Tachycardia With 1:1 Atrioventricular Ratio:

Fetal tachycardia is defined as a fetal heart rate >180 bpm. If the heart rate is >180 bpm with a 1:1 atrioventricular ratio, SVT is the probable diagnosis. In cases of persistent sinus tachycardia, long-acting thyroid-stimulating receptor antibodies from maternal Graves' disease should be considered as acause of fetal hyperthyroidism.

Fetal Tachycardia With >1:1 or <1:1 Atrioventricular Ratio:

Atrial flutter is the most common primary fetal atrial tachycardia, characterized by atrial rates >300 bpm with 2:1, 3:1, or 4:1 AV conduction. Atrial ectopic tachycardia may also cause fetal tachycardia with variable AV block. If the AV ratio is <1, the rhythm is likely ventricular tachycardia (VT) or junctional ectopic tachycardia (JET), which are associated with AV dissociation, retrograde venous flow, and rapid hydrops even at ventricular rates <180 bpm. In suspected VT or JET, maternal anti-Ro/SSA antibodies (extended panel) should be tested, the fetal heart evaluated for structural lesions, and long QT syndrome (LQTS) considered—especially before using QT-prolonging drugs such as amiodarone or sotalol. Often maternal lupus is associated with enodocardial fibroelastotic changes.

Fetal Bradycardia With 1:1 Atrioventricular Ratio:

Fetal bradycardia is defined as a sustained fetal heart rate below the third percentile for gestational age. Bradycardia with a 1:1 AV relationship may result from an ectopic atrial rhythm associated with congenital heart defects, sinus rhythm in inherited arrhythmias such as LQTS, or secondary causes including maternal anti-Ro/SSA antibodies, viral infections, or maternal medications. Parental ECGs and fetal or neonatal genetic testing may identify underlying inherited variants.

Fetal Bradycardia With >1:1 Atrioventricular Ratio:

Fetal bradycardia with an AV ratio >1:1 can result from advanced second- or third-degree AV block (AVB) or blocked atrial bigeminy. High-grade AVB may be associated with maternal anti-Ro/SSA antibodies, LQTS, left atrial isomerism, or congenitally corrected transposition of the great arteries, with poor prognosis associated with structural heart defects. Anti-Ro/SSA antibodies cause AVB in 2–6% of positive mothers, recur in 18% of subsequent pregnancies, and can also lead to endocardial fibroelastosis, valvular involvement, and dilated cardiomyopathy, worsening outcomes even without AVB (1). Prolonged isovolumic relaxation time is now considered as a predictor of fetal LQTS and should also be evaluated. Distinguishing second-degree AVB from blocked atrial bigeminy relies on atrial interval assessment: second-degree AVB shows regular intervals, while blocked atrial bigeminy has variable intervals and may lead to SVT. Early detection via handheld Doppler and prompt treatment within 12 hours is critical for improving fetal outcomes.

Management:

The primary objective of anti-arrhythmic therapy in the fetus is to restore and maintain a normal heart rate, prevent or reverse cardiac dysfunction, reduce the risk of developing endocardial fibroelastosis, and avoid premature delivery along with its associated complications. A comprehensive clinical evaluation should be undertaken, supported by basic laboratory investigations such as a full blood count, thyroid function tests, and assessment of serum electrolytes. It is essential to determine whether the arrhythmia is paroxysmal or incessant and to assess for signs of haemodynamic compromise. These may include cardiomegaly, atrioventricular valve regurgitation, ventricular dysfunction, and/or fetal hydrops. Identifying these features is crucial for prognosis and guiding management decisions.

Prenatal transplacental therapy is the preferred management strategy when there is evidence of fetal haemodynamic compromise. Delivering a premature infant with hydrops and tachyarrhythmia poses a significant clinical challenge and carries a high risk of neonatal mortality. In the absence of fetal hydrops, treatment is successful in approximately 90% of cases. However, when hydrops is present, the success rate decreases to around 60% (5).

Early management strategies for fetal tachyarrhythmias included digoxin, which showed some clinical benefit. However, it was later recognised that in the presence of fetal hydrops, transplacental transfer of digoxin is significantly reduced, limiting its effectiveness. Currently, flecainide and sotalol are the preferred anti-arrhythmic agents. Both have been shown to be widely effective, generally well tolerated, and associated with a low risk of in-utero demise. Notably, flecainide has demonstrated greater efficacy than digoxin, particularly in cases complicated by fetal hydrops (6).

Fetal management of SVT and atrial flutter and immune-mediated high-degree AV block have been specified by Batra *et. al*. and briefly summarised in tables 2 and 3 (1).

Transplacental therapy:

Prenatal therapy for fetal arrhythmias can be administered either directly to the fetus or indirectly via the mother. Direct fetal therapy involves injecting medications into the fetal peritoneal cavity under echocardiographic guidance, a procedure that requires specialized expertise and is not widely available. In contrast, transplacental therapy using maternal antiarrhythmics is more convenient, as treatment can be initiated in an inpatient setting. Before starting maternal antiarrhythmic therapy, a baseline 12-lead ECG is essential to exclude conditions such as Wolff-Parkinson-White syndrome, prolonged QT interval, or myocarditis, which could increase the risk of maternal complications. Our unit recommends serial maternal ECG monitoring is to detect possible side effects, and serum levels of digoxin, flecainide, and, if feasible, sotalol to be measured at appropriate intervals to ensure therapeutic efficacy and prevent toxicity. Flecainide and sotalol require baseline ECG assessment prior to initiation, while digoxin can be administered intramuscularly in cases where the fetus has a poor biophysical profile or hydrops that is refractory to transplacental treatment. This approach allows effective management of fetal supraventricular tachycardia while prioritizing maternal safety.

Outcomes:

Fetal tachyarrhythmias can be successfully treated in up to 90% of cases when the arrhythmia is uncomplicated. The need for long-term postnatal antiarrhythmic therapy is generally limited, and long-term outcomes are typically favourable. In contrast, the prognosis for fetal ventricular tachycardia is less favourable, as it is often associated with underlying myocardial pathology or conditions such as LQTS.

Outcomes for fetal bradyarrhythmias have historically been less satisfactory compared with tachyarrhythmias. However, treatment of third-degree AVB with corticosteroids, with or without intravenous immunoglobulin (IVIG), has been reported to improve survival rates significantly. Reported fetal, neonatal, and one-year survival rates are approximately 95%, 93%, and 89%, respectively. These results support a more optimistic and proactive management approach (1).

A team-based, multidisciplinary approach—including fetal medicine specialists, fetal and pediatric cardiologists, and neonatologists—has markedly improved outcomes. The timing of delivery should be determined collaboratively within this team. In the absence of fetal compromise or other obstetric concerns, an early-term delivery should be considered.

Conclusion:

Current evidence for transplacental treatment is still evolving. However, outcomes have improved significantly with the involvement of specialist fetal cardiologists and appropriate timing of delivery. In the absence of poor prognostic indicators—such as fetal hydrops—a favorable outcome can generally be expected when adequate rate control is achieved

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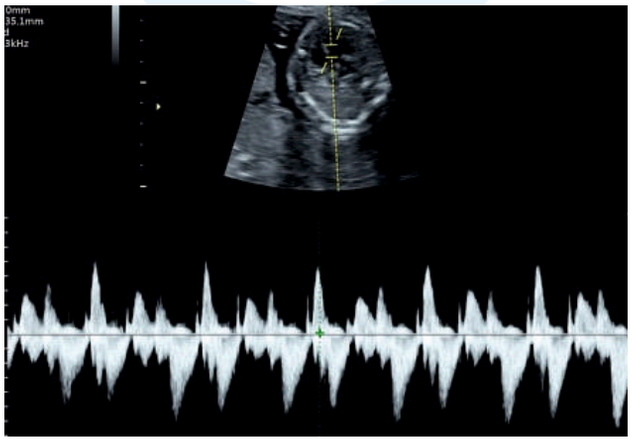

Figure 1. Alternate conducted premature atrial ectopic beats and normal beat, resulting in pause and irregular cardiac rhythm

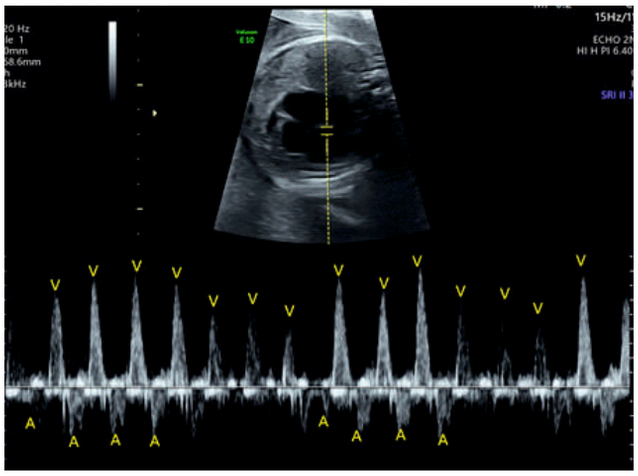

Figure 2. Fetal LVOT Doppler showing multiple ventricular premature beats, not preceded by atrial activity

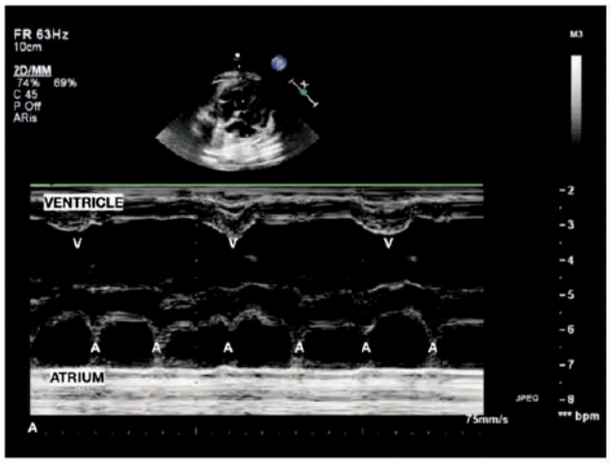

Figure 3. Complete fetal AV block. Atrial contractions occuring regularly and independently of ventricular contractions. The M-mode cursor is positioned to simultaneously transect the atrial and ventricular walls, allowing visualization of both atrial (A) and ventricular (V) contractions. [Adapted from 4]

Table 1. Causes of Fetal arrhythmias (adapted from 1)

Atrioventricular ratio	Bradycardia: below third percentile for gestational age	Tachycardia >180 bpm	Irregular rhythm
1:1	Sinus bradycardia (Maternal Anti -Ro, Maternal viral infection, Ectopic atrial rhythm, Inherited arrhythmia) Ectopic atrial rhythm	Sinus tachycardia (Maternal Trab, Maternal stimulants, Maternal Anti -Ro) SVT (AVRT, PJRT) Some VT and JET AET	PAC, PVC without retrograde ventriculoatrial conduction, Intermittent SVT
>1:1	AVB (Maternal anti -Ro, Kearns -Sayre syndrome, Long-QT syndrome) BAB	Atrial flutter (AET)	Type 1, second - degree AVB Intermittent type 2, second - degree AVB Nonconducted PACs PVC with retrograde ventriculoarterial conduction
<1:1	Ventricular bigeminy	VT, JET	Intermittent VT or JET

AET indicates atrial ectopic tachycardia; AVB, atrioventricular block; AVRT, atrioventricular reentry tachycardia; BAB, blocked atrial bigeminy; JET, junctional ectopic tachycardia; PAC, premature atrial contractions; PJRT, permanent junctional reciprocating tachycardia; PVC, premature ventricular contractions; SVT, supraventricular tachycardia; TRab, thyroid-stimulating receptor antibody; and VT, ventricular tachycardia

Table 2. Approach to management of Fetal SVT and AFL (adapted from 1)

Condition	Hydrops Status	Line	Treatment Options
SVT	No Hydrops	1st Line	Flecainide
		2nd Line	Flecainide + Digoxin OR Sotalol + Digoxin
		3rd Line	Flecainide + Sotalol OR Amiodarone
	Hydrops	1st Line	Flecainide + Digoxin
		2nd Line	Sotalol + Digoxin OR Flecainide + Sotalol
		3rd Line	Amiodarone ± Digoxin ± Direct Treatment
AFL	No Hydrops	1st Line	Sotalol
		2nd Line	Sotalol + Digoxin
		3rd Line	Sotalol + Flecainide OR Amiodarone
	Hydrops	1st Line	Sotalol + Digoxin
		2nd Line	Sotalol + Flecainide OR Amiodarone
		3rd Line	Amiodarone + Digoxin ± Direct Treatment

AFL indicates atrial flutter; and SVT, supraventricular tachycardia

Table 3. Approach to perinatal management of immune-mediated high-degree AVB (adapted from 1).

	Medications	Criteria	Treatment / Action
1	Dexamethasone	Continue to birth if anti-Ro+	Start 8 mg/day → 4 mg/day after 2 weeks → 2 mg/day after 28 weeks
2	IVIG	If EFE or incomplete AVB	1 g/kg (max 70 g) every 3–4 weeks
3	β-mimetic	If VR < 50 bpm	Salbutamol 10 mg in 3 doses **or** Terbutaline 10–30 mg in 4 doses

AVB indicates atrioventricular block; EFE, endocardial fibroelastosis; IVIG, intravenous immunoglobulin; and VR, ventricular rate in beats per minute.

Medications need to be upgraded if the line of management is unable to control fetal AVB.

Weekly assessment by Obstetrics + Fetal cardiology is required followed by delivery at tertiary care center.

Dive Deer

SOCIETY OF FETAL MEDICINE

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Arrhythmias Beyond the ECG: Genetic Counseling in Fetal Rhythm Disorders

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Background the rapid heartbeat observed on the fetal doppler or ultrasound brings a sense of relief and joy, both to the parents and the consultant physician. However, 1-3% of every pregnancy may be detected with fetal arrhythmia. Fetal arrhythmias may be caused due to different factors and may also have genetic etiology. Genetic factors may include chromosomal anomalies, copy number alterations and significant variant(s)/ mutation(s) in clinically relevant genes. Genetic fetal arrhythmia is usually severe and may result in miscarriages, still births or even live births of babies with inherited arrhythmia syndromes. Understanding the etiology of fetal arrhythmia can help in management and counseling of the family.

Introduction

Inherited fetal arrhythmias are cardiac rhythm anomalies of the fetal heart caused by genetic changes that affect cardiac ion channels or structural heart proteins. Unlike transient rhythm disturbances, these arrhythmias arise from heritable cardiac disorders, most commonly channelopathies, and may present during pregnancy as persistent bradycardia, tachycardia, or irregular heart rhythms.

With the innovation of Doppler echocardiography and fetal ECG, the detection of fetal arrhythmias has increased with higher sensitivity. It is of clinical importance in the prenatal setting because they lead to:

•Hydrops fetalis

•Intrauterine demise (IUD)

•Neonatal Sudden Cardiac Death

•Lifelong arrhythmia risk

This is especially helpful to prepare families regarding their ongoing pregnancy for the management antenatally and post birth. It is important to evaluate such cases with a detailed family history to identify the possibility of an inherited arrhythmia syndrome. Genetic counseling plays an important role in identifying such fetal rhythm disorders, detecting genetic changes, recurrence risk in subsequent pregnancies, prevention and family screening.

Different Types of Fetal Arrhythmias [1]

I.Extrasystoles

ii.Fetal Tachyarrhythmias

iii.Bradyarrhythmias

Inherited Rhythm Disorders [2]

Arrhythmias may be caused due to cardiomyopathies, or cardiac ion channel anomalies. Cardiac Channelopathies are genetic or acquired conditions wherein the ion channels (such as the sodium, potassium and calcium) have a significant dysfunction.

1. Long QT Syndrome (LQTS) [3]

LQTS is a cardiac channelopathy which is caused due to genetic alterations in around 17 genes including KCNQ1, KCNH2 and SCN5A genes which account for almost 90% of cases. LQTS is characterized by a long QT interval followed by “T”- wave abnormality in ECG. In adults it may be manifested with clinical symptoms such as seizures, sudden syncope, unexplained death, pathological QTc during stress or exercise or with specific triggers. However, in certain cases it may not show any clinical manifestation. The prenatal diagnosis of LQTS is often challenging but may manifest as abnormalities in fetal heart rate, in particular fetal bradycardia, hydrops fetalis and may also lead to intrauterine fetal demise (IUFD).

2. Short QT Syndrome (SQTS)

SQTS is marked by accelerated repolarization that leads to extremely short QT intervals followed by tall, peaked T-waves on the ECG. This channelopathy is caused due to gain of function mutations in genes encoding the potassium channels- KCNH2, KCNQ1, and KCNJ2. These genes are usually inherited in the autosomal dominant pattern. The most common manifestation is sudden cardiac arrest which is most seen to manifest between 14-40 years of age. It may present as atrial fibrillation in neonates and young children. It is often challenging to capture SQTS in the prenatal setting and it thus important to investigate families with cardiac histories for a potential risk in their future pregnancies

3. Brugada Syndrome (BrS) [4]

Brugada Syndrome a potentially life-threatening cardiac disorder which predisposes young, otherwise healthy individuals to fatal ventricular arrhythmias and sudden cardiac death. The condition is most caused by autosomal dominant mutations in the SCN5A gene, which encodes a cardiac sodium channel, although other genetic and environmental factors may modulate disease expression. These mutations impair sodium current, creating conduction abnormalities and predisposing to reentrant arrhythmias. Brugada Syndrome is not commonly detected during the prenatal period; however, it is important to screen a mother predisposed to this syndrome and the fetus who may be at risk.

4. Catecholaminergic polymorphic ventricular tachycardia (CPVT) [5]

CPVT is an arrhythmia characterized by episodic syncope occurring during exercise. The underlying cause of such episodes is due to fast ventricular tachycardia. In some cases the arrhythmias may resolve spontaneously or in other cases ventricular tachycardia may degenerate into ventricular fibrillation and cause sudden death. The mean age of onset of symptoms is usually 7 to 12 years of age and may manifest as late as the fourth decade of life. This disorder is usually inherited in an autosomal dominant pattern (RYR2, CALM1, CALM2, CALM3, CASQ2, KCNJ2) or in the autosomal recessive pattern (CASQ2, TECRL, TRDN).

Genetic Evaluation of Inherited Rhythm Disorders in the Fetal Medicine Clinic

Inherited Arrhythmias are usually caused due to clinically significant variants in ion channels genes of the heart or sometimes due to certain cardiomyopathies. Most of these disorders are first identified postnatally and may have very generalised symptoms. Some of these disorders are lethal and the first symptom at onset may be sudden cardiac arrest. Thus, it is important to evaluate a family with a known history of cardiac problems. In the fetal medicine clinic, it is important to gauge the risk of both mother and fetus being of having an inherited arrhythmia syndrome. Thus, genetic counseling plays an important role in identifying patients and their fetuses who may be at risk, conducting a genetic evaluation and discussing the management options during the prenatal and/ or post-natal period.

Clinical Evaluation.

It is important to evaluate the current pregnancy, fetal cardiac parameters and maternal cardiac parameters during the period of gestation. Any fluctuations or anomalies captured of Fetal Doppler or Fetal ECG should be followed up and evaluated. In cases of persistent arrhythmias, hydrops fetalis, IUD it is important to conduct genetic evaluation of the cardiac genes comprising genes responsible for channelopathies, cardiomyopathies and other relevant genes. However, detailed family history and genetic counseling is recommended for such identified cases.

Genetic Counseling.

In the Pre-test Genetic Counseling, it is important to understand the clinical history of the pregnancy in question. Are there any current clinical symptoms such as bradyarrhythmia or tachyarrhythmia, increasing edema/ hydrops fetalis or IUD? It is important to take detailed family history to assess the risk of inherited cardiac condition including arrhythmias and other channelopathies. These details include family history of individuals with:

i.Existing heart conditions
ii.Addition of pacemakers
iii.History of sudden cardiac arrest
iv.History of sudden death
v.History of antenatal cardiac history, IUDs, hydrops fetalis

In case the current gestation is affected, it is important to test the fetal sample or abortus material to identify clinically significant variant(s)/ mutation(s) in causative genes associated with the cardiac phenotype. In many cases the parental samples are also simultaneously tested to help in clinical correlation and variant / mutation segregation.

In cases where there is a significant family history without a clinical manifestation in the current gestation, it is important to test the parents to identify if they are carriers / silent carriers of clinically significant variant(s)/ mutation(s) in channelopathy genes. Most of the inherited arrhythmias show incomplete penetrance which leads to partial symptoms in mutation carriers or may sometime not manifest in certain generations. It is important to identify these mutations to not only identify the risk in the current fetus but also for the medical management of the parent with the identified mutation.

Test Recommendation

The recommended test is dependent upon the clinical and family history of the case.

a.In certain cases where there are multiple systems affected and may show syndromic characteristics in the fetus, a chromosomal microarray or karyotype may be recommended on the fetal sample.

b.In cases where there is a cardiac phenotype noted in the clinical history of the fetus, DNA based NGS tests such as Clinical Exome Sequencing or Whole Exome Sequencing may be recommended on the fetal sample. In certain cases a Trio NGS test may be recommended for simultaneous testing of the fetal sample and parental samples

c.In cases with a strong family history without fetal cardiac phenotype, parental couple carrier testing using NGS may be recommended based upon the history assessed. Upon identification of a variant/ mutation with a likely risk to the fetus, further fetal sample testing may be recommended after genetic counseling

Post-Test Genetic Counseling and Family Screening

The identification of a mutation in either the affected fetus or in the parents assists in identifying the cardiac disorder likely present in the family. It helps estimate the risk of recurrence in subsequent pregnancies. In an ongoing pregnancy, it can help prepare the family for the possible outcomes of the fetus and options of management antenatally or postnatally (if available). It is also important in identifying the parent who may be at risk of having the disorder but may not have manifested symptoms. It assists in timely intervention. Further, identification of such a familial mutation also helps identify other people in the family who may be at risk of having inherited the same mutation leading to a risk of channelopathies / arrhythmia. Thus, family screening is important for timely intervention and management especially for inherited arrhythmia syndromes where there may be general symptoms at onset or may lead to sudden cardiac arrest.

Conclusion

Genetic Counseling plays a key role in risk assessment, family screening and perinatal management of fetal arrhythmia syndromes. Early identification of an at-risk family or pregnancy can help in evaluation, risk estimation and guidance on management options of the pregnancy and maternal health. Identification of such clinically significant variants can further detect other at-risk family members and can prevent sudden cardiac death among many generations. Such a multidisciplinary approach including maternal fetal medicine, pediatric cardiology and genetics is crucial in the detection and management of different inherited fetal rhythm disorders.

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Dive Deeper

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Identification of Time-sensitive Lesions and Referral Pathway

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Introduction one of the primary diagnostic objectives of antenatal ultrasound (USG) in the modern era is identification of critical congenital heart diseases (CCHD) during fetal life, which are often time-sensitive. This allows for a planned transition from the parallel fetal circulation to the series-based neonatal circulation. Failure to identify these conditions prenatally often leads to sudden deterioration of the neonate as the physiological shunts—the ductus arteriosus and foramen ovale—close shortly after birth. These time-sensitive or critical congenital heart diseases(CCHD) require procedural intervention within the first hours or days of life to prevent cardiovascular collapse. Fetal arrhythmias are also time-sensitive and may lead to similar hemodynamic compromise due to heart failure, but we will discuss structural heart problems only in this article.

Classification

Classification of CCHD is fundamentally linked to the concept of ductal dependency and the necessity for adequate mixing between parallel circulations. But there are some non duct-dependent lesions with potential for antenatal or immediate postnatal complication for different reasons. These lesions may be categorised into:

A. Duct-dependent pulmonary blood flow: Pulmonary atresia (PA) with intact ventricular septum (IVS), Critical pulmonary stenosis (PS), Tetralogy of Fallot (ToF) with PA/critical PS, Severe Ebstein's anomaly with functional PA

B. Duct-dependent systemic blood flow: Hypoplastic left heart syndrome(HLHS), Interrupted aortic arch, Critical Coarctation of Aorta (CoA) or aortic stenosis (AS)

C. Mixing-dependent lesions: Transposition of great arteries (TGA) with IVS

D. Obstructive pulmonary venous lesions: Total anomalous pulmonary venous return (TAPVR), obstructed

E. Unique circular shunt: Severe Ebstein's anomaly with tricuspid regurgitation (TR) allows reversed ductal flow back to the right ventricle (RV), leading to low cardiac output and related complications with a high risk of fetal demise.

F. Miscellaneous: Tetralogy of Fallot with absent pulmonary valve, Truncus arteriosus with severe truncal valve regurgitation, Atrioventricular septal defect with severe AV valve regurgitation etc..

Identification

The efficacy of managing critical cardiac lesions is dependent upon the timing and accuracy of prenatal diagnosis. National and international guidelines, including those from the International Society of Ultrasound in Obstetrics and Gynaecology (ISUOG) and the American Society of Echocardiography (ASE), emphasise specific windows for screening and the necessity of specialised evaluation for high-risk pregnancies.

The optimal period for transabdominal fetal cardiac screening is between 18 and 22 weeks of gestation. At this stage, cardiac structures have achieved sufficient size for detailed visualisation of the four-chamber view (4CV), the outflow tracts, and the three-vessel trachea view (3VT). Utilising high-frequency probes and optimised system settings, such as high frame rates (above 40 Hz) and low persistence, is critical for good imaging and identifying subtle anomalies.

Comprehensive screening must include:

1. Fetal Situs: Assessment of laterality is the first step, ensuring the stomach and heart are correctly positioned on the left side of the fetus.

2. Four-Chamber View: Evaluation of chamber size and symmetry, movement of AV valves, crux of heart, and ventricular function.

3. Outflow Tract Views: Verification of origin and crossover of great arteries.

4. Three Vessel Trachea View: Interrogation of relative sizes and arrangements of the superior vena cava (SVC), ductal (DA) and aortic (AA) arches.

Advances in ultrasound technology have enabled the detection of major cardiac defects as early as 12 to 14 weeks. However, first-trimester echocardiography exhibits lower sensitivity compared to mid-trimester scans. Consequently, any cardiac evaluation performed in the first trimester must be repeated during the standard 18–22 week window.

Some lesions, notably Hypoplastic Left Heart Syndrome (HLHS) and Coarctation of Aorta (CoA), are "evolutionary" and may not be fully manifest until the late second or even the third trimester. HLHS often begins as critical aortic stenosis. As gestation progresses, the high afterload and reduced flow to the left ventricle inhibit its growth, eventually leading to the classic hypoplastic phenotype.

Echocardiographic predictors of such progression include:

1. Aortic flow velocity: > 2 m/s

2. Aortic arch flow: Retrograde flow in transverse arch

3. Foramen ovale flow: Reversed, left to right
4. Mitral inflow: Monophonic inflow pattern
5. Pulmonary venous flow: Abnormal/reversed flow

The identification of these markers has led to the development of fetal cardiac intervention like aortic valvotomy. The goal is to re-establish forward flow, decompress the left ventricle, and encourage its growth, thereby preserving the possibility of a biventricular repair postnatally.

Identification also relies on hemodynamic risk stratification.Ultrasound technology is used to identify fetuses at high risk for acute cardiorespiratory instability (ACRI) within the first two hours of birth. Key markers include:

*** Restrictive Foramen Ovale (RFO):**

For hypoplastic left heart syndrome (HLHS), RFO is predicted by an foramen vale (FO) diameter < 4 mm (bigger in TGA, smaller in normal heart). A fixed or hypermobile FO flap also suggest RFO. Size of FO has also been compared with other cardiac structures to get a standard ratio for restrictive physiology. By Doppler evaluation, RFO could also be identified by right to left flow doppler velocity >40 cm/s.

• Premature Ductus Arteriosus Constriction:

The ductus arteriosus should remain widely patent throughout fetal life. Premature constriction, often idiopathic or secondary to maternal NSAID use, increases right ventricular afterload, leading to right heart enlargement, tricuspid regurgitation and potential hydrops. In fetuses with TGA, ductal constriction is particularly dangerous because it limits the volume of oxygenated blood reaching the right heart, exacerbating the effects of an RFO and significantly increasing the risk of early neonatal death.

*** Ductal Dependency:**

Identified by retrograde flow in the transverse aortic arch or ascending aorta, which suggests critical left-sided obstruction. Similarly, retrograde flow in ductal arch suggests critical obstruction or atresia of right ventricular outflow.

*** Cardiovascular Profile (CVP) Score:**

A 10-point echocardiographic assessment used to evaluate cardiovascular well-being in high-risk pregnancies. The CVPS typically evaluates five parameters, with 2 points awarded for each normal finding (total of 10), and points deducted for abnormalities. The parameters are: Heart size, Ventricular function, Hydrops, Venous doppler and Arterial doppler.

Referral Pathway

Identification of a suspected cardiac anomaly or a high-risk factor necessitates fetal echocardiography assessment at the earliest and referral to a paediatric cardiologist. After proper assessment, the level of care (LOC) is planned based on anticipated hemodynamic stability. Such a proposed care system may be like this:

• LOC 1 (Low Risk): Minor shunt lesions (e.g., small VSD). Delivery can be conducted at a community hospital.

• LOC 2 (Low-Moderate Risk): Stable TOF or large septal defects. Delivery at a regional hospital with a Level II/III NICU.

• LOC 3 (Moderate Risk): Suspected coarctation, pulmonary atresia or stable TGA. Delivery at a tertiary centre with on-site paediatric cardiology facility and PGE1 availability.

• LOC 4 (High Risk): TGA or HLHS with restrictive atrial septum or obstructed TAPVR. These cases require delivery at a dedicated paediatric cardiac centre with immediate access to catheterisation lab and cardiac surgery.

High-acuity cases (LOC 3 and 4) require the involvement of multidisciplinary team,including pediatric cardiologists, obstetricians, neonatologists and cardiac surgeons, to coordinate delivery and immediate postnatal care.

Conclusion

Timely identification and referral, proper counselling of prospective family and involvement of multidisciplinary team are essential for delivery planning and ideal care of these time-sensitive critical congenital heart diseases.

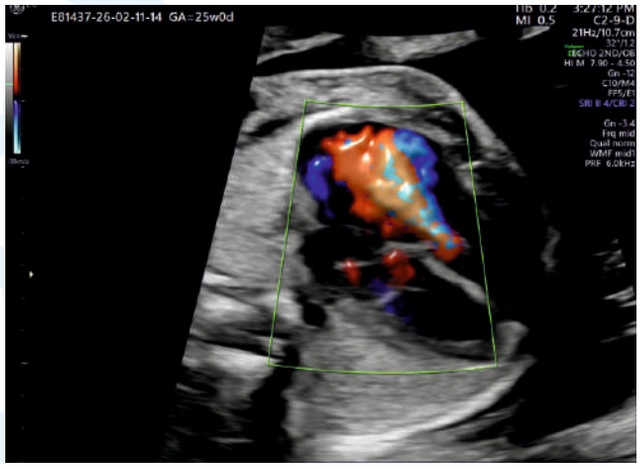


Fig 1- Ebstein's Anomaly: Cardiomegaly with hugely dilated RA and severe TR

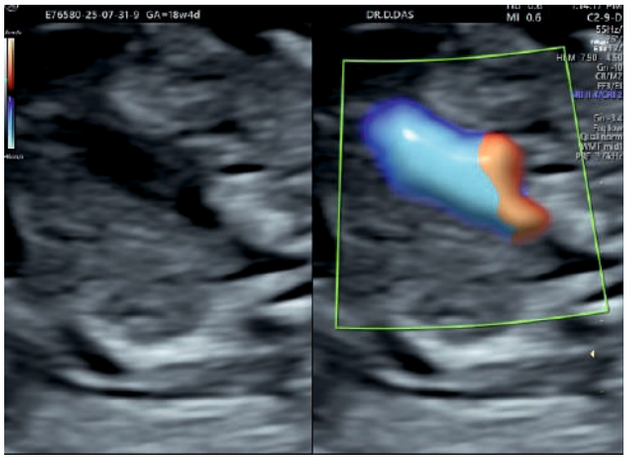
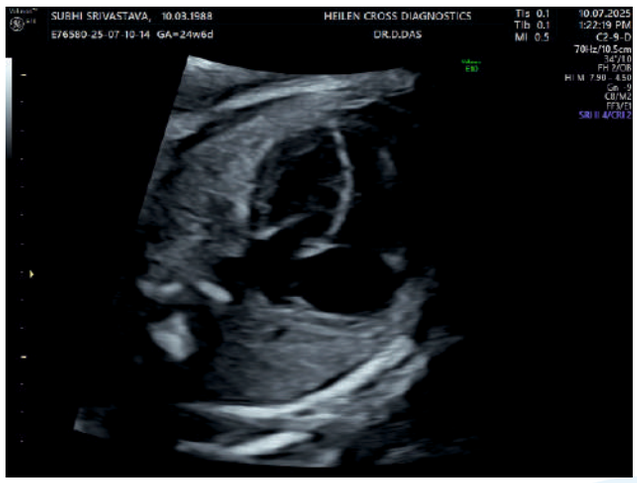


Fig 2- HLHS: Hypoplastic ductal arch with reversed flow



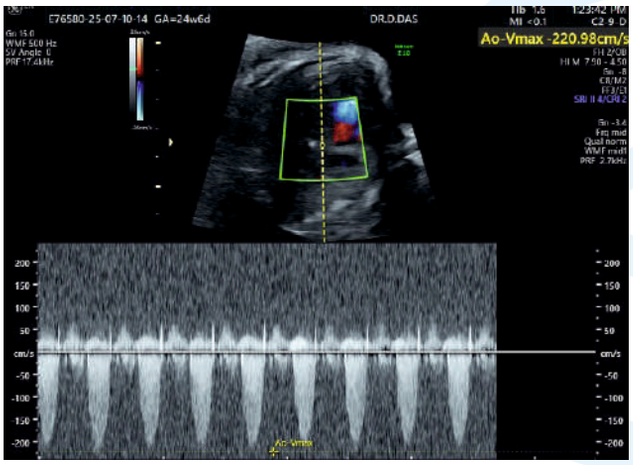


Fig 3 & 4 - Developing HLHS: Deceptive normal looking 4 CH view, but LV dysfunction with increased velocity across aortic valve (>2 m/s) suggest early intervention

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SOCIETY OF FETAL MEDICINE

Genetic Counseling in Congenital Heart Defects:
From Isolated CHD to Syndromes

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Introduction Congenital heart defects (CHDs) affect 8–12 per 1,000 live births and are the leading cause of birth defect-related infant mortality. Their etiology spans chromosomal aneuploidy, copy number variants (CNVs), monogenic disorders, and multifactorial inheritance. Approximately 20–30% of CHDs occur as part of a recognizable genetic syndrome, with the remainder considered 'isolated' — though even these carry heritable risk. As genetic counselors, our role is to translate genomic complexity into actionable family guidance across three domains: syndromic recognition, testing strategy, and recurrence risk counseling.

1. Isolated vs. Syndromic CHD

Isolated CHDs — including ventricular septal defect (VSD), atrial septal defect (ASD), pulmonary stenosis, and bicuspid aortic valve — occur without major extracardiac anomalies. Despite the label, molecular analysis often reveals pathogenic variants in cardiac transcription factors (NKX2-5, GATA4, TBX5) or signaling genes (NOTCH1, JAG1), reflecting an oligogenic or multifactorial architecture.

Syndromic CHDs, by contrast, form part of a broader multi-system phenotype. Identifying a syndrome refines prognosis, guides surveillance, and clarifies inheritance. Key examples are summarized in Table 1.

Syndrome	Genetic Cause	Common Cardiac Lesion(s)
Down Syndrome (T21)	Trisomy 21	AVSD, ASD, VSD
Turner Syndrome (45,X)	Monosomy X	Bicuspid aortic valve, CoA
22q11.2 Deletion Syndrome	22q11.2 deletion	Conotruncal defects (TOF, IAA-B, TA)
Noonan Syndrome	RASopathy (PTPN11 etc.)	Pulmonary stenosis, HCM, ASD
Williams Syndrome	7q11.23 deletion	Supravalvular aortic stenosis
Holt-Oram Syndrome	TBX5 (AD)	ASD, VSD, conduction defects
CHARGE Syndrome	CHD7 (AD)	Conotruncal defects, AVSD

Table 1. Selected syndromes associated with CHD. AD = Autosomal Dominant; AVSD = Atrioventricular Septal Defect; CoA = Coarctation of Aorta; HCM = Hypertrophic Cardiomyopathy; IAA-B = Interrupted Aortic Arch Type B; TA = Truncus Arteriosus; TOF = Tetralogy of Fallot.

2.When to Suspect Syndromic CHD

A structured clinical assessment should guide suspicion for an underlying genetic syndrome. Key red flags include:

Cardiac Features:

– Complex or conotruncal anatomy: Tetralogy of Fallot, truncus arteriosus, interrupted aortic arch type B, and double-outlet right ventricle carry a substantially elevated rate of chromosomal or monogenic etiology compared to simple shunt lesions.

– CHD with cardiomyopathy: The combination of structural and functional disease raises suspicion for RASopathies, storage disorders, or mitochondrial conditions.

– Aortic or great vessel anomalies: May indicate heritable connective tissue disorders (Marfan, Loeys-Dietz) or Turner syndrome.

Extracardiac Features:

– Dysmorphic facial features, even subtle — any recognizable gestalt warrants referral

– Intellectual disability, developmental delay, or neurodevelopmental concerns

– Multiple congenital anomalies across organ systems

– Thymic hypoplasia or hypocalcemia (22q11.2); lymphedema or webbed neck (Turner); hearing loss or coloboma (CHARGE)

Family and Obstetric History:

– First-degree relative with CHD, especially the same lesion type

– Recurrent pregnancy loss, advanced maternal age, or consanguinity

– Abnormal prenatal screen: nuchal translucency ≥3.5 mm, abnormal cfDNA, or hydrops fetalis

> **Clinical Pearl**
> Any three of the following should prompt comprehensive genetic evaluation: complex CHD + one extracardiac anomaly + positive family history + abnormal neurodevelopment. This 'rule of three' applies regardless of the specific cardiac diagnosis.

Testing should follow a stepwise, phenotype-driven approach to maximize diagnostic yield while managing cost and interpretive burden

– Chromosomal Microarray (CMA): Current standard-of-care for CHD with extracardiac features. Detects CNVs — including the common 22q11.2 deletion — at far higher resolution than karyotype. Recommended as first-tier in neonates with syndromic-appearing CHD.

– Karyotype: Remains first-line when clinical features strongly suggest aneuploidy (Down, Turner, Edwards syndrome), and detects balanced translocations missed by array.

– Targeted gene testing / panels: When phenotype suggests a specific Mendelian syndrome (e.g., PTPN11 for Noonan; TBX5 for Holt-Oram; FBN1 for Marfan), single-gene or panel-based NGS is efficient and cost-effective. Counsel families on the

possibility of variants of uncertain significance (VUS).
– Exome / Genome Sequencing (WES/WGS): Increasingly first-tier in complex or unexplained CHD. Trio analysis (proband + parents) significantly increases yield byidentifying de novo variants, which account for 8–10% of isolated CHD and a higher proportion of syndromic cases. Diagnostic yield: ~10–29% in isolated CHD; ~40–50% with extracardiac features. Pre-test counseling must address secondary findings per ACMG SF v3.2 guidelines.

4. Recurrence Risk Counseling

When a Genetic Cause is identified:
– Autosomal Dominant (AD): 50% recurrence risk per pregnancy. Expressivity is often variable — an affected parent with a mild phenotype can have a child with severe CHD. This must be communicated explicitly.
– Autosomal Recessive (AR): 25% recurrence risk for carrier parents. Relevant for consanguineous families and rare metabolic CHD syndromes.
– Chromosomal aneuploidy: Trisomy 21 recurrence is ~1% above age-related background risk for mothers under 40. Parental karyotyping is essential when a translocation is identified.
– De novo variants: Parental recurrence risk is low (<1–2%), primarily reflecting gonadal mosaicism. Siblings carry a slightly elevated but still low risk (~1–2%).
– Inherited CNVs: If inherited from an apparently unaffected parent, siblings have a 50% chance of inheriting the CNV, but penetrance may be incomplete — a nuance requiring careful counselling.

Multifactorial/Isolated CHD- Empiric Risks:

When no causative variant is found, empiric population data guide counselling:

Situation	Empiric Risk	Notes
One affected sibling (parents unaffected)	~2–4%	*Varies by lesion type*
Two affected siblings	~10%	*Reconsider Mendelian cause*
Affected mother	~4–6%	*Higher than paternal transmission*
Affected father	~2–3%	
Left-sided lesions (HLHS, CoA, BAV)	~8–12%	*Familial clustering well-documented*

Table 2. Empiric recurrence risks for multifactorial CHD. Lesion-concordance (same defect recurring) is observed in ~50% of familial cases. BAV = Bicuspid Aortic Valve; CoA = Coarctation of Aorta; HLHS = Hypoplastic Left Heart Syndrome.

Prenatal Options:

–Fetal echocardiography: Recommended at 18–22 weeks for all pregnancies at elevated risk. Sensitivity ~85–90% in high-risk populations.
– CVS or amniocentesis: Provides definitive fetal genotyping when a chromosomal or monogenic cause is known.
– Preimplantation genetic testing (PGT-M): Available for families with a confirmed Mendelian CHD syndrome.
– Cell-free fetal DNA (NIPT): Screens for common aneuploidies; expanded panels may detect some large CNVs (e.g., 22q11.2), though sensitivity for microdeletions varies and a negative result is not a guarantee.

Conclusion.

The genetic landscape of CHD is complex but clarifying rapidly. A structured approach — identifying syndromic red flags, applying stepwise genomic testing, and delivering individualized recurrence risk counseling — equips families to make informed decisions and empowers clinicians to optimize care. As genotype-phenotype relationships continue to be refined, collaboration between cardiologists, geneticists, and genetic counselors remains essential to translating genomic discovery into meaningful clinical benefit.

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SOCIETY OF FETAL MEDICINE

FETAL CARDIAC GENETICS

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Congenital heart disease (CHD) is the most common birth defect affecting nearly 10 to 12 per 1000 live born infants (1%–1.2%). CHD represents an important cause of morbidity and mortality during infancy and childhood, and nearly 25% of affected infants have critical CHD requiring intervention within the first year of life.

The etiology of CHD is complex and heterogeneous, involving the interaction of genetic susceptibility and environmental factors. Over the past two decades, improvements in prenatal ultrasound imaging, fetal echocardiography, and genomic testing technologies have dramatically increased the prenatal detection of congenital cardiac anomalies.

Genetic factors are estimated to contribute to approximately 20–30% of congenital heart defects, particularly when extracardiac anomalies are present.

Understanding the genetic basis of fetal cardiac defects is crucial for:

- Determining recurrence risks
- Guiding prenatal counseling
- Identifying associated syndromes
- Planning perinatal management.

Approach to fetal cardiac defects

Fetal cardiac anomalies can present as an isolated problem or can be associated with other system problems . Once a cardiac defect is identified, a detailed search for associated anomalies is essential.

Maternal history

- Maternal medical disorders (e.g., diabetes mellitus, autoimmune disease)
- Exposure to teratogenic medications
- Infections during pregnancy
- Mode of conception, including assisted reproductive technologies

Obstetric history

- Previous pregnancy losses
- History of congenital anomalies
- Previous child with congenital heart disease

Family history

- Construction of a three-generation pedigree
- Assessment for familial cardiac disease
- Consanguinity

Evaluation of previous affected child

If a previous child has CHD, documentation should include:

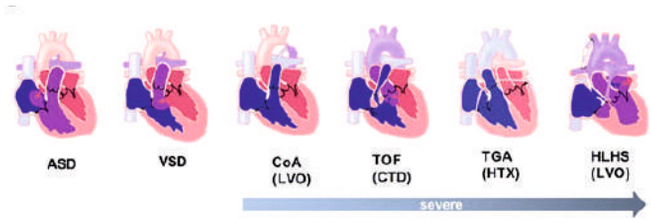
- Type of cardiac lesion
- Surgical interventions performed
- Developmental outcomes
- Syndromic features

Parental examination may identify subtle features of genetic syndromes such as skeletal abnormalities or dysmorphism that may suggest inherited conditions. Based on the clinical phenotype and family history, appropriate genetic testing strategies and counseling can be planned.

Spectrum of Congenital Cardiac Defects

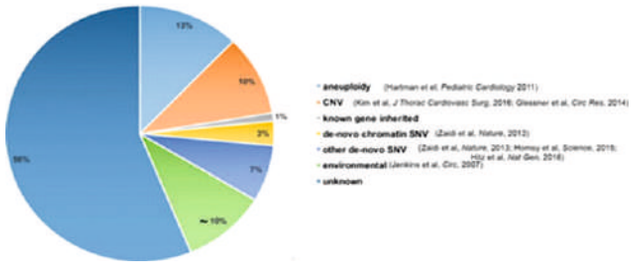
Congenital cardiac defects represent a broad spectrum of abnormalities ranging from simple septal defects to complex structural malformations.

Ventricular septal defect remains the most common congenital heart defect and accounts for approximately 30–40% of all CHD cases.



Genetic Etiology of Congenital Heart Disease

1. Chromosomal
2. Single gene mutations
3. Multifactorial
4. Mitochondrial



Chromosomal problems and fetal cardiac defects:

Chromosomal problems can be of 3 types

1. Numerical aberrations – Trisomy,Monosomy,Triploidy
2. Structural aberrations – Translocations,Deletions,Inversions
3. Microdeletions – also called copy number variants

When to suspect chromosomal problems
When a fetus presents with
Increased nuchal translucency with a structural heart defect like Venticular septal defect- peri membranous or muscular,endocardial cushion defect, outflow tract anomalies, valvular dysplasia, Hypoplastic cardiac chambers, coarctation of aorta or a complex cardiac anomaly.

Common aneuploidies and cardiac anomalies:

• Trisomy – 21- Approximately 40–50% of fetuses with trisomy 21 have congenital heart disease. ASD,VSD,PDA,AVSD Complex cyanotic heart disease

• Trisomy 18 – Cardiac anomalies occur in more than 90% of affected fetuses and include: Septal defect , Shunts,hypoplastic right heart, valvular defects

• Trisomy 13 - Shunts, hypoplastic ventricles , conotruncal defects

• Turners syndrome – coarctation of aorta, bicuspid aortic valve complex cyanotic heart disease

• Triplody – Shunts, hypoplastic right heart

Genetic counselling will vary according to maternal age, mode of conception, maternal risk factors, order of pregnancy, mode of conception etc.

-For lethal anomalies parents may choose to discontinue pregnancy but importance of genetic testing and autopsy should be emphasised . The information got through these tests will help plan next pregnancy better.

-If it is a surgically correctable abnormality a multidisciplinary counselling with obstetrician, fetal medicine consultant, cardiologist, cardiac surgeon, neonatologist and geneticist is arranged so that couple understand what it takes to go through the process – emotional and financial implications have to be explained in detail. Genetic testing and ensuring normalcy is crucial for correctable cardiac abnormalities.

Prenatal screening for chromosomal abnormalities includes:

- Combined first-trimester screening
- Non-invasive prenatal testing (NIPT).

However, these are screening tests and not diagnostic.

When a structural fetal anomaly is detected, invasive diagnostic testing is recommended.

Prenatal Invasive Procedure:

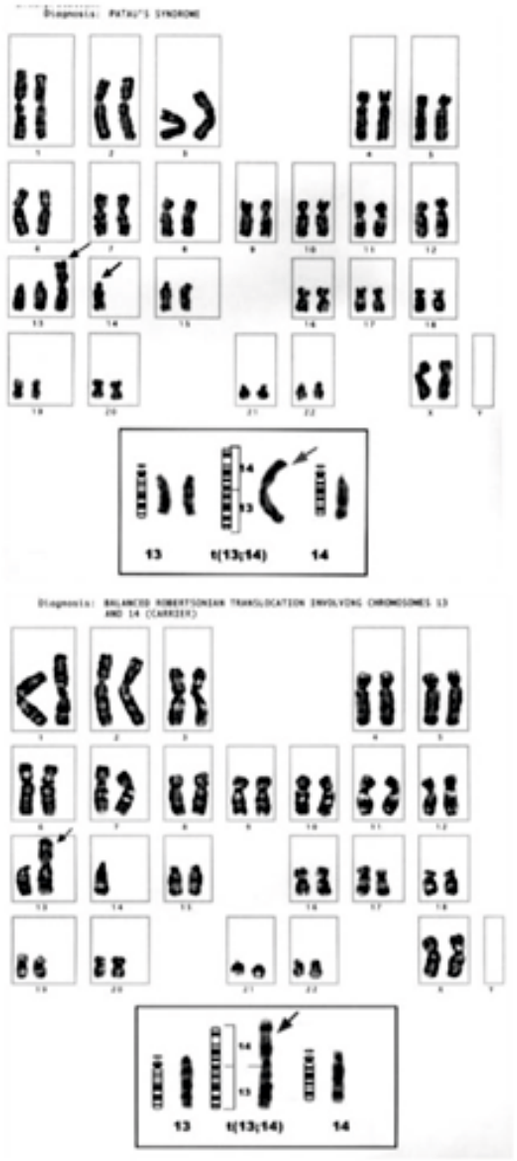
If gestational age is 11-14 weeks we could do a CVS

If gestational age is > 16 weeks we could do an amniocentesis

Test of choice:

Primi,non consanguineous, no significant personal or family history with isolated cardiac anomaly from above group : QFPCR (rapid aneuploidy screening)to rule out common aneuploidy and if negative we can proceed with chromosomal microarray. Chromosomal microarray analysis is now recommended as the first-tier diagnostic test for fetus with structural anomalies, as it can detect clinically significant copy number variants not identifiable by conventional karyotyping

Example of a structural chromosomal abnormality.



Fetus with unbalanced 13/14 Translocation

Mother with balanced 13/14 translocation

A fetus presented with a peri membranous VSD, polydactyly, dysmorphic facies – So an amnio was done which revealed unbalanced translocation 13,14 chromosomes. Parental karyotype revealed –Mother is a balanced carrier of 13/14 translocation .Recurrence risk is 10-15 % in every pregnancy.

So the reproductive choices would be:

- Prenatal testing with Karyotype & microarray in every pregnancy
- Donor gamete
- Preimplantation genetic testing PGT-SR

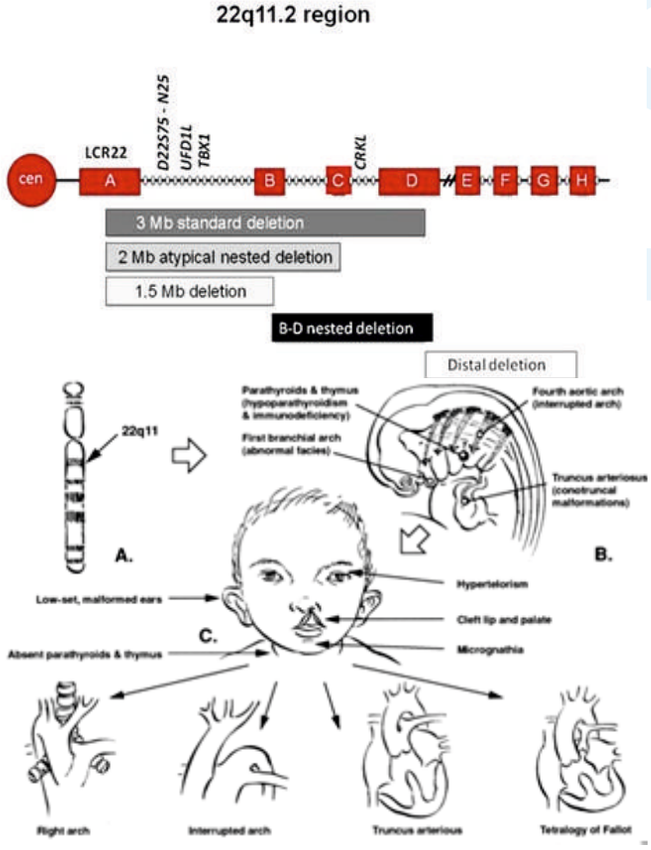
Microdeletion in fetus:

Approximately 3% to 10% of isolated CHD cases are caused by CNVs

Case example:

Digeorge syndrome

The major clinical manifestations of 22q11.2DS include conotruncal malformations (ventricular septal defect, tetralogy of Fallot, interrupted aortic arch, and truncus arteriosus), palatal abnormalities (velopharyngeal incompetence, submucosal cleft palate, bifid uvula, and cleft palate), immune deficiency, characteristic facial features, and learning difficulties. Hearing loss can be sensorineural and/or conductive. Laryngotracheoesophageal, gastrointestinal, ophthalmologic, central nervous system, skeletal, and genitourinary anomalies also occur. Psychiatric illness and autoimmune disorders are more common in individuals with 22q11.2DS



In 22q11. 2DS caused by a 3.0 (2.54)-Mb deletion. Inheritance patterns can be varied

--Denovo in more than 90% of individuals.

--Inherited from a heterozygous parent in about 10% of individuals.

--60% of 22q11.2DS caused by an atypical or nested 22q11.2 deletion inherited the deletion from an affected parent.

--Offspring of affected individuals have a 50% chance of inheriting the 22q11.2 deletion.

Once the 22q11.2 deletion has been identified in an affected family member, prenatal testing using MLPA, or Chromosomal Microarray studies for a pregnancy at increased risk and preimplantation testing are possible.

• Cardiac defect	• Estimated22q deletion frequency %
• Interrupted aortic arch	• 50-89
• VSD	• 10
• With Normal arch	• 3
• With aortic arch anomaly	• 45
• Truncus arteriosus	• 34-41
• Tetralogy of Fallot	• 8-35
• Isolated arch anomalies	• 24
• DORV	• <5
• TGA	• <1

Other microdeletion syndromes associated with CHD include:

- Williams syndrome (7q11.23)
- Cri-du-chat syndrome (5p deletion)
- Wolf–Hirschhorn syndrome (4p deletion)

Genetic Counseling for microdeletion:

If a pathogenic microdeletion is found in the child – we should see the breakpoints of deletion or duplication,see if it is pathogenic or VOUS (till date there is no cientific evidence to say it is disease causing)the number of genes located in that place and expected phenotype.

Parents are checked if variant in the child is confirmed pathogenic

If one parent has the CNV recurrence is 50% in every pregnancy

If parents are normal – recurrence is low

However germline mosaicism should be noted

Single Gene disorders and fetal cardiac defects:

Single gene mutations affecting cardiac developmental pathways include genes involved in:

- Chromatin remodeling
- Notch signaling
- Cilia function
- Sarcomere structure
- RAS signaling pathways.

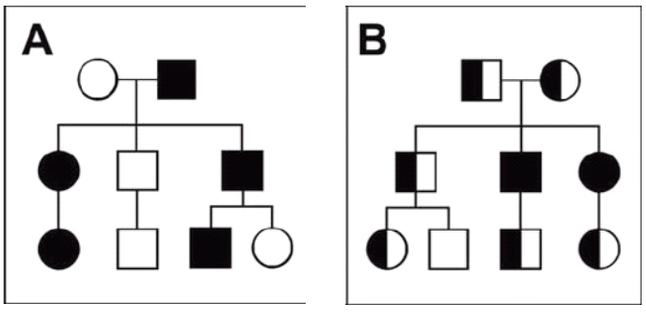
Monogenic causes are estimated to account for approximately 10–20% of severe congenital heart defects:

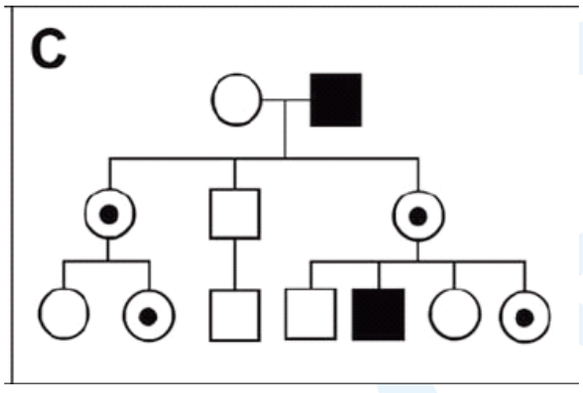
- Providing a molecular diagnosis not only has implications for psychosocial well-being and future family planning but can also significantly influence the clinical management of some patients with variants in specific genes known to be associated with the future development of conduction defects or cardiomyopathies. The inheritance can be autosomal dominant,autosomal recessive or X linked recessive inheritance.

Important genes implicated include:

- NKX2-5
- TBX5
- TBX20
- GATA4.

Presentation of single gene disorders are usually cardiomyopathy or syndromes. Isolated cardiac defects can be because of mutation in cardiac transcription genes like NKX,GATA4 etc These are generally isolated & non syndromic





Autosomal dominant	Autosomal recessive	X linked recessive
Males & females affected	Males & females affected	Males affected
Variable severity	Parents are carriers	Females carriers
50% recurrence risk if parents transmit Low–if denovo However germline mosaicism to be kept in mind	25% recurrence risk	50% risk of males to be affected 50% females– carriers

Examples of Autosomal dominant fetal cardiac defects:
Tuberous sclerosis, Marfan syndrome ,cardiomyopathy, HoltOram syndrome

Examples of Autosomal recessive:
Cardiomyopathy (metabolic –Pompe,or non metabolic)syndromes like Ellis van creveld syndrome(AVSD)Short rib polydactyly syndrome

Examples of X linked recessive:
Cardiomyopathy(Fabry.Duchenne muscular dystrophy, Barth syndrome)

Genetic Testing for Single Gene Disorders:
Next-generation sequencing techniques such as whole exome sequencing (WES) are increasingly used to identify monogenic causes of CHD.

Whole exome sequencing is recommended when:
- Chromosomal microarray is normal
- Complex cardiac defects are present
- Extracardiac anomalies are identified
- There is a strong suspicion of a monogenic disorder

Diagnostic yield is particularly high in:
- Heterotaxy syndrome
- Cardiomyopathy
- Syndromic CHD
- Familial recurrence of CHD

Variants are classified according to ACMG guidelines as:
- Pathogenic
- Likely pathogenic
- Variant of uncertain significance
- Benign.

Prenatal testing should generally be offered only for variants classified as pathogenic or likely pathogenic

Mitochondrial Disorders:
Mitochondrial disorders may arise due to mutations in:
- Mitochondrial DNA
- Nuclear genes involved in mitochondrial function.

Mitochondrial mutations are transmitted exclusively through the maternal lineage. All her males & females will be affected. Disease severity depends on the degree of heteroplasmy, making prediction of clinical severity difficult. Extent of severity & organ involvement will depend on % of abnormal mitochondria in a particular organ.
So prenatal testing is best avoided as we cannot predict severity of the disease. We monito carefully by scans.

Multifactorial inheritance:
Many cases of CHD arise from multifactorial inheritance, involving interactions between genetic predisposition and environmental influences. Empiric risk assessment is given to such patients – and genetic tests will be essentially normal.

- I child with CHD – Rec.risk 1.5 to 5 %
- If 2 children with CHD - Risk 5 to 10%
- If mother has CHD – Risk for child -2.5 to 18%
- If father has CHD- Risk for child 1.5 to 3%

Left outflow obstructions have a higher rate of recurrence than other heart defects.

Genetic Counseling and Reproductive Options:
Recurrence risk summary:
Chromosomal –
- Aneuploidy - <1% - no need to test parents
- Structural – Test parents – If one is carrier -10-15% recurrence
- Microdeletion- Test parents – If one is positive -50% recurrence
- Single gene disorders – Test parents
- AD – 50% recurrence (variable penetrance & expressivity)
- AR – 25 % recurrence risk
- XLR – 50 % risk of getting affected sons;50% risk of getting carrier daughters (test Mother alone)

Genetic Evaluation in Fetal Congenital Heart Disease:

Cardiac Lesion–Specific Genetic Associations and Recommended Genetic Tests

Cardiac Lesion	Common Genetic Associations	Recommended Genetic Test
Atrioventricular septal defect (AVSD)	Trisomy 21	QF-PCR / Karyotype + Chromosomal microarray
Conotruncal defects (TOF, Truncus arteriosus, Interrupted aortic arch)	22q11.2 deletion syndrome	Chromosomal microarray ± targeted 22q11 testing
Coarctation of aorta / Left-sided obstructive lesions	Turner syndrome, familial left heart lesions	QF-PCR / Karyotype + Microarray
Cardiac rhabdomyoma	Tuberous sclerosis (TSC1, TSC2)	Targeted gene testing / Exome sequencing
Heterotaxy syndrome	Cilia-related gene mutations	Whole exome sequencing
Familial septal defects	NKX2-5, GATA4 mutations	Gene panel / Whole exome sequencing
Cardiomyopathy in fetus	Sarcomere gene mutations, metabolic disorders	Whole exome sequencing

Suggested Genetic Testing Algorithm for Fetal Congenital Heart Disease:

Step 1: Detailed fetal echocardiography and targeted anomaly scan

↓

Step 2: Assess for extracardiac anomalies and obtain detailed family history

↓

Step 3: Offer invasive testing (CVS or amniocentesis)

↓

Step 4: Rapid aneuploidy testing (QF-PCR / FISH)

↓

Step 5: Chromosomal microarray analysis (first-line genomic test). If there is a high suspicion of monogenic disorder – Whole exome sequencing – first line genetic test

↓

Step 6: If microarray is normal and suspicion remains → Whole exome sequencing

↓

Step 7: Parental segregation testing and genetic counseling based on results

Conclusion:

Fetal cardiac genetic disorders are common. If identified we need to take history, pedigree ,examine previous affected child and screen parents. Genetic tests are planned according to phenotype. Genetic counselling is useful to understand course of the disease, recurrence risk, interpretation of results and reproductive option suggestions

SOCIETY OF FETAL MEDICINE

Ethical Dilemma and Counselling After Diagnosis of CHD By Fetal Echocardiography

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Fetal echocardiogram has been into tremendous advancement in the last two decades due to technological advancements and fetal cardiac imaging. Initially, it was an investigation of choice, and was advised for a select group of pregnant ladies fulfilling the indications. However, with time, fetal echocardiogram is being used more as a screening tool in a major chunk of pregnancies, to rule out any fetal cardiac structural anomalies and rhythm issues – which may jeopardise the growth and development of the baby not only in utero, but after birth as well.

The incidence of congenital heart diseases is about 8 – 10 per thousand live births in the general population and approximately 77% of major cardiac defects can be detected by first-trimester ultrasound screening. In the United States, where CHD occurs in about 1% of births, most cases are diagnosed prenatally.

A fetal echo which is usually done between 16 – 18 weeks in many countries, is done in about 20 – 22 weeks in our practice. It is useful in early detection, formation of an opinion amongst the parents, the mental preparation to face the clinical situation, arranging for the birth in a cardiac centre and the economics involved in the subsequent interventions or surgery – if required, which may often require multi stage operations / interventions. Furthermore, increased maternal stress may affect the growth and/or neurocognitive development of the fetus. Rise in maternal cortisol levels due to stress may influence fetal development.

In a country like ours, where economics plays a pivotal role in each and every bit of life, planning a baby and ushering in a new life needs exquisite and elaborate thoughts on the part of the parents and extended family. Every extra cost cuts a corner in the pocket of the family, even amongst the wealthy middle class, leave alone the families with average income. Though the cardiac treatment of the baby may be carried out with government subsidies in some cases, it does take some time for help to reach. In some cases, the help may not suffice for multistage operations, interventions and the medical management in between the stages. Hence couples would dream for an ideal situation, where the baby is born in the medical facility and the parents walk away with a smile and the baby in their arms a couple of days from the baby's birth. Often complications are not on the radar, but the associated costs are also not apprehended. Medical insurances, though promised – deny reimbursements in congenital matters, specially in congenital heart diseases. The government often fail to verbalise in those matters due to unknown reasons.

Parents are often apprehensive about the quality and the quantity of life the treatment can provide. In the modern era, almost every congenital heart disease can be treated with medical, interventional and surgical procedures, barring only a select few with very high operative risks – such as HLHS (Hypoplastic Left Heart Syndrome). Today's cardiac surgeries and interventions are highly sophisticated , along with the state of the art imaging and navigation facilities , with the post operative care being featured with the latest gadgets, we can boast of the superb results of cardiac surgery and interventions – which ranges between 2 – 5 % mortality rates in most centres for the standard operations or interventions.

The operated children have a near normal growth pattern, mental acumen and can enjoy life as it is almost similar to their normal peers – barring a few. The "quality" and "quantity "of life can both be comparable. Even then, the amount of mental trauma and agony the news of an unborn child with a CHD bears on the minds of the parents – is unthinkable. The economics, social stigma, maternal guilt with fear of ostracization (the world is still a male dominated society) all get added to the fear of losing the child at any point of time. The helpless feeling of NOT being able to give birth to a "perfect life" lingers on for a lifetime. Sometimes one parent might be supportive to the idea of ushering in a life with defects, the other parent and family members may not agree to the idea and would prefer a "perfect child" , a concept which probably doesn't exist. Even if there are no defects in the heart, can there be any guarantee that the other organs won't spring in any surprise diseases later in life ? Hence, many parents choose NOT to usher in the child in the world would be the best option available, even if counselled well by the caregivers. Hence the decision to "let go" may not be well taken by many, and the deep wound and guilt remains. In an era where the general fertility rates are dropping with more and more ladies choosing career over family in the reproductive age groups which coincides with the peak of their career – the birth of a baby is a major event with a career break. More so, in the era of IVF and ovum banking, how many chances can we take before creating a perfect baby – isn't it a food for thought?

More so , if the diagnosis of the fetus with CHD is made late in pregnancy beyond the time limit for a MTP from the government , it is difficult to console the parents that the rules don't allow to terminate after 24 weeks of pregnancy , even if the unborn child has a deadly disease which can make its life endangered or the parents do not wish to carry the burden of such a child and want to break free looking forward to a fresh chance of creation. Such permissions are to be taken from the legal system which is often difficult and time consuming.

All said and done, what are the options then? With a high fertility rate in our country, we can always argue that for the parents, it is perhaps easier to give up on this child and try for a "more perfect child" later? On a rather philosophical note – is it even possible that as humans we play God and create perfection at each and every attempt? Can we take over nature? Are there any guarantees that the next child will be perfect throughout life? Even though the newborn may be apparently normal at birth, can't there be any genetic disorders which may manifest later? Are we all NOT subjective to any malignancies or a serious disorder which may manifest a little later in life, or even can an accidental death be predicted? Obviously, we the mere mortals can anything but predict these events accurately.

Parental counselling for fetal heart disease is complex and multidimensional. Significant expertise in fetal cardiology and physiology, potential progression of CHD, postnatal treatment strategies and knowledge of long-term sequelae is necessary. A structured approach, together with continuous improvement of communicative skills, may lead to more effective counseling for parents following a diagnosis of CHD in the fetus. The social, cultural and religious backgrounds should be respected as well during the decision.

There are different recommendations by international societies on counseling after diagnosis of fetal CHD. The American Heart Association (AHA), The Association for European Paediatric and Congenital Cardiology (AEPC), have published their own guidelines while the International Society of Ultrasound in Obstetrics & Gynecology (ISUOG) have important statements on the association between CHD neurodevelopmental delay and its impact on prenatal counselling.

Ideally parents should be counseled shortly after the fetal echocardiogram – preferably in their vernacular language , in simple non medical terms as much as possible. We usually use hand drawn diagrams – though others may use models or online materials to explain the heart disease and therapeutic options. The team of counsellors should include a pediatric cardiologist , fetal medicine specialist, the obstetrician , a neonatologist and a geneticist and the meeting should be held in private, in a separate counselling room. In an Indian scenario, sometimes extended family members may request to be present in the counselling session – the decision to include them in the meeting rests upon the medical team and the need of the hour. The opinion of cardiac surgeons may be sought if the parents desire to talk to the surgical team , regarding the prospects of surgery and the prognosis. A non-directive approach should be obtained – thereby providing neutral, evidence-based information without any personal bias or allowing parental, societal, or medical pressures to dictate the decision.

ISUOG has reiterated the importance of neurodevelopmental issues of children diagnosed with CHD. While the majority of neonates do well, some may have neurological problems in the long-term, the exact category of which cannot be predicted prenatally. Children with univentricular hearts eg. HLHS, are at increased risk of neurodevelopmental delay, which should always be mentioned during counseling. It has to be reiterated that with co-morbidities such as abnormalities in other organs (a common association) or with genetic anomalies the risk may even be higher. Possible associated extra-cardiac malformations may include many organ systems: renal (agenesis, dysplasia) and gastrointestinal disease, abdominal wall defects, spine defects, and more. Associated genetic diseases include numerical chromosomal abnormalities, single gene disorders and genetic syndromes. The risk for additional genetic diseases may vary from < 1% in simple Transposition of the Great Arteries up to 65% in Atrioventricular septal defects. Apart from the typical chromosomal abnormalities (Trisomy 13, 18 and 21) , single gene disorders exist like Noonan syndrome , Holt-Oram and Alagille syndrome. In CHDs with conotruncal anomalies counseling should include the microdeletion syndrome DiGeorge/22q11, as it for its wide clinical variety and multidisciplinary counseling.

Potential diagnostic limitations must be discussed with parents. These may occur due to an unfavorable fetal position, maternal habitus, oligo- or polyhydramnios, or gestational age. Some defects which may be missed are: VSD , conotruncal anomalies, coarctation of aorta , Tetralogy of Fallot, AV septal defect , Aortic valve stenosis despite utmost care. It should be counselled during the initial assessment that despite utmost care in viewing the fetal heart, optimal imaging may not be obtained due to several reasons, leading to a miss in the diagnosis. The idea of a fetal echo is not only to pick up the defects so as to plan and strategize the delivery and subsequent therapy, but also to prepare the prospective parents with a receptive mental condition, so as the diagnosis shouldn't come as a surprise. However, if a diagnosis is missed, utmost care should be ascertained so as any wrong doings should be nullified by helping the parents towards a complete solution.

After initial counselling, some parents may request written documents – which they should be provided. In the age of internet and social media , the parents may already be informed about the disease and the treatment options. However , it is advised that the learning should be from authentic websites , and not the ones which provide incomplete information – thus creating a confusional stage.

Fetal cardiac interventions (FCI) are still not in routine use at all centres. RCTs have been performed and are incorporated into practice in some fetal therapies like endoscopic laser therapy for twin-to-twin transfusion syndrome (TTTS), or intrauterine surgery for myelomeningocele. FCI are mainly performed for critical aortic stenosis with evolving HLHS, critical pulmonary stenosis / pulmonary atresia with intact ventricular septum and in HLHS with a restrictive foramen ovale – only in centers who have perfected the art with dedicated teams. These centres have shown survival advantage and more biventricular repairs have been shown for critical aortic stenosis with evolving HLHS. However, the risks of intrauterine death and preterm delivery loom large and these interventions are still controversial.

These procedures may best be done at select referral centres. And the pros and cons may be discussed with the parents during counselling. Several arrhythmias in the fetus are successfully treated by administering the antiarrhythmic medication onto the mother at a higher dosage, so that the required amount may cross the placenta and reach the fetus. This method of therapy has a high success rate and should routinely be offered to the parents if the need be. Usually a pediatric cardiologist supervises the treatment along with regular fetal echoes to monitor the fetal heart rate and the well being of the fetus , to rule out development of any pericardial effusion / hydrops fetalis.

In summary , the parents should be offered an impartial view about not only the condition of the heart, but the associated conditions and genetics as well – as per the merit of the case. They should be counselled in the first sitting , preferably in their vernacular language, with visual and authentic informative aids, by a team of doctors comprising of pediatric cardiologists, fetal medicine specialists, obstetricians, geneticists, social workers and pediatric cardiac surgeons, as and when required. It should be an unbiased view, but the socio economic, cultural and religious views and above all, the mother's health and parental views should be the final verdict.

Leaving alone the parental side or even the newborn's cause, what really happens in the minds of the fetal cardiologist while on the judge's chair? Whatever said and done, how would we as doctors judge ourselves that we have counselled the parents well and above all, delivered an impartial judgement to the yet unborn child, who cannot speak but has equal rights to a dignified life? Have we passed the right judgement or have we snatched that right in the pretext of perfection? The answer is perhaps blowing in the wind.

Dive Deeper

SOCIETY OF FETAL MEDICINE

Science and Stories– A Quarter to Remember
Quarterly Highlights for Bengal SFM

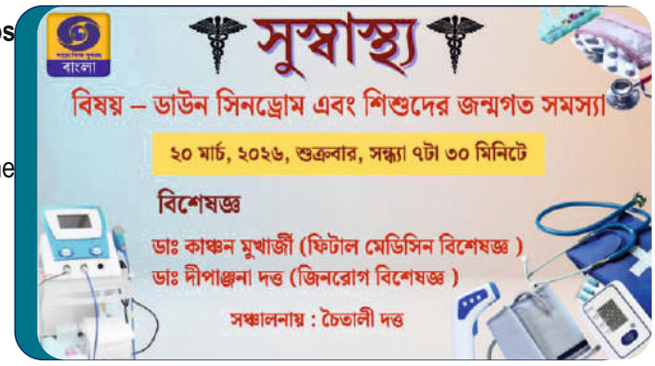
Building Knowledge, Expanding Reach
Over the past quarter, **SFM Bengal** has taken significant strides in advancing both **professional education** and **public awareness** in fetal medicine and genetics. Through focused outreach and collaborative efforts, we continue to strengthen the knowledge ecosystem across West Bengal.

Academic Outreach & Capacity Building
East Midnapur District Programme
SFM Bengal conducted a highly engaging academic outreach programme in East Midnapur, featuring esteemed experts:
D**r. Kamal Oswal, Dr. Kushagradhi Ghosh, Dr. Kanchan Mukherjee, Dr. Prasanna Roy, and Dr. Dipanjana Datta.**

A key highlight was an in-depth session on USG **abnormalities and Haemoglobinopathies** which was received with **great enthusiasm** by participating clinicians, reflecting the growing interest in advanced genetic diagnostics.

Malda Seminar
A large-scale seminar held in Malda witnessed **participation from over 100 doctors**, marking it as a major success.
The session was led by:
Dr. Shankar De (Secretary, SFM Bengal), Dr. Vinayak Das, and Dr. Dipanjana Datta
The discussions focused on integrating **fetal medicine and genetics into clinical practice**, with excellent engagement from attendees.

Public Awareness Initiatives
SFM Bengal remains committed to spreading awareness at the community level:
• **Bengali Educational Brochures** developed on **thalassemia and sickle cell disease** to improve accessibility and understanding among the general population.
• **Awareness Video Campaign** created by SFM Bengal and **broadcast acros district and urban health centres,** ensuring wide outreach.



World Down Syndrome Day – Expanding Voices
This year, SFM Bengal elevated its outreach efforts for World Down Syndrome Day:
• **Radio Outreach:**
Dr. Kanchan Mukhopadhyay and Dr. Dipanjana Datta were featured on **Akashvani Kolkata,** enabling outreach to even the **most remote regions, including the Sundarbans.**
The initiative received **widespread appreciation** from diverse communities.
• **Awareness Programmes:**
Both experts also actively participated in public engagement programmes, promoting **awareness, inclusion, and early diagnosis** in Bengal Door darshan

Looking Ahead
This quarter reflects SFM Bengal's continued commitment to:
• **Academic excellence**
• **Collaborative learning**
• **Inclusive public health awareness**
We look forward to expanding our impact further in the coming months.

SOCIETY OF
FETAL MEDICINE

Society of Fetal Medicine
Northeast Chapter
Annual Conference 2026

10th - 12th April

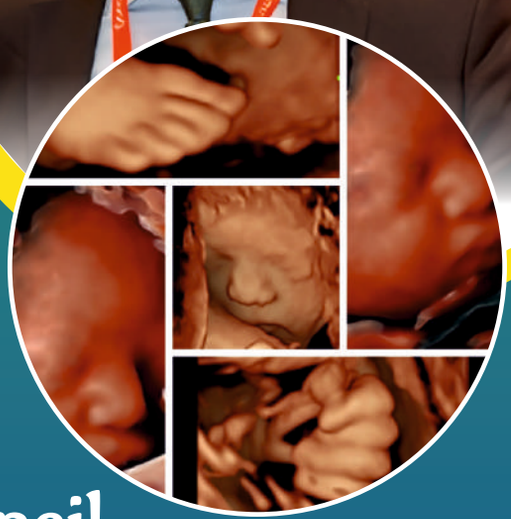
Hotel Radisson Blu, Guwahati, Assam, India

Everyday Approach to Fetal Anomalies

For the first time
in the Northeast
The Grand Master in Person

Prof. Rabih Chaoui

Happy to Announce



Awarded 6 Credit Points From Assam Medical Council

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www.societyoffetalmedicine.org

Conferences International

SOCIETY OF FETAL MEDICINE

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Bengal Chapter
Annual Conference 2026

Block Your Date

Sunday, 5th July, 2026
Kolkata, West Bengal

Registration will Start Soon

For Any Query:
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Conferences International

Dear Fetal Heart Watchers,

Reflecting. Debating. Arguing.

Occasionally agreeing, though we try not to make a habit of it.

Through SamVaad, the yearly congregation of the SFM think-tanks, the Society has been quietly doing something rather remarkable, designing the future of fetal medicine in India.

On the other hand, the author's home team, the Bengal Chapter, deserves a heartfelt ovation (pun entirely intended, and unapologetically so). Producing these newsletters with such regularity is no small feat. It requires persistence, patience, and the rare ability to chase deadlines that seem to have a mind of their own. Nevertheless, these efforts go a long way in elevating shared learning.

Well, conflict of interest disclosed and respectfully parked aside, let us proceed to business now.

Across clinics, practitioners remain deeply immersed in their familiar rituals: freezing the perfect 4-chamber view, tracing the outflow tracts with almost meditative precision, aligning septa, and engaging in quiet internal debates about whether that tiny echogenic focus is a villain in disguise or merely an innocent bystander enjoying undue attention.

All this careful scrutiny is directed toward a remarkable biological structure, the fetal heart. A tiny masterpiece of engineering that begins its tireless work long before most families feel ready to announce the pregnancy on social media. This issue of the newsletter, quite fittingly, is devoted entirely to that small but profoundly consequential organ.

Fetal cardiology, however, is not merely about capturing perfect images or achieving elegant cuts. It is, in many ways, about carrying responsibility. The task does not end with anatomical description. A physician inevitably steps into a family's emotional ecosystem, a delicate space where science meets uncertainty, where optimism negotiates with fear, and where decisions are not always comfortable companions. For all these, one needs a heart large enough to accommodate much more than pure anatomy.

To those practitioners who have chosen this path, take a bow. It is no ordinary work. Every careful scan, every thoughtful conversation, every teaching session becomes a quiet investment in a future that none of us will fully witness. Appreciation is due to those brave-hearts who zoom in, slow down, question, teach, doubt, and still show up the next day to do it all over again.

This issue also arrives at a time when the world outside the ultrasound room feels unusually heavy. In West Asia, conflict continues to unfold, leaving behind loss, displacement, and lives irreversibly altered. Behind every headline are families, parents and children, whose futures have been abruptly rewritten. For many, the gentle rhythm of everyday life, the very rhythm that medicine strives to protect, has been replaced by uncertainty. It is difficult not to think of the many unborn children who will begin life under the shadow of such turmoil, their first stories already shaped by forces far beyond their control.

Yet perhaps that is precisely why our work must continue.

Let us carry on, quietly and steadily, in a world that often feels fragile and unpredictable.
Let the rhythm continue.
Let the sound sustain

Truly yours
In fetal frequency,
The Fetus Uncle
(Dr Kanchan Mukherjee)

Where Science Meets Satire