Basic Sonographic Evaluation of Fetal Heart in A Common Congenital Abnormalities: Review

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Abstract

Congenital heart diseases (CHD) are considered as anomaly of the heart that has been seen after birth. They are classified according to abnormalities in the walls; great vessels and valves .These abnormalities affect the normal flow of blood. Sonographic screening during 2nd trimester of pregnancy could detect the defects .Nowadays great improvement has been achieved with the use of advanced ultrasound machine and good training for the operator.
Keywords: ASD, CHD, Ebstein's anomaly, TGA, TOF and VSD.

التقييم السونوغرافي الاساسي لقلب الجنين في حالات الشذوذ الخلقي الشائعة

زينة مهدي الهنداوي, الاء سعد العتابي

الخلاصة

الموجات الفوق الصوتية الاساسية لتقيم قلب الجنين في التشوهات الخلقية الشائعة المرض القلبي الخلقي هو عبارة عن شذوذ في القلب يمكن ملاحظته بعد الولادة ويمكن تصنيفه الى تشوهات في الجدار تشوهات في الاوعية الدموية الكبرى وتشوهات الصمام التي تؤثر على تدفق الدم الطبيعي فحص الموجات الفوق الصوتية خلال الفصل الثاني من الحمل يمكن الكشف عن العيب والتي تحسنت في الوقت الحالي مع وجود اجهزة الموجات الفوق الصوتية والتدريب الجيد للمشغلين على هذه الاجهزة

Introduction

Congenital heart defects (CHDs) are the most frequent congenital malformations, the costliest hospital admissions for structural defects and the leading cause of infant general and malformations related mortality.Most cases of CHDs occurred without knowing the risk factors, and its detection is strongly depending on examiner experience. There are many ultrasound findings seen in screening of early gestation that give a clue and raise suspicion to the presence of CHD. These finding are : 1:Thickened nuchal translucency. 2: Abnormal cardiac axis. 3: Abnormal reverse flow in ductus venosus. 4: Tricuspid regurgitation. 5: Gross extracardiac fetal malformation. 6: Early fetal hydrops [1].

Thickened Nuchal Translucency (NT): NT thickness measurement can be performed between 11^{th} – 13^{th} week gestation which considered a good method in establishment of fetal chromosomal abnormalities [2] and also associated with elevated incidence of heart defects [2, 3]. In the beginning "study of Hyett *et al.* [3], it was observed that 56% of fetuses with major CHD present with a thickened NT and the authors suggested utilizing the NT measurement as a mean for CHD screening as well. The mechanism that connects the presence of thickened NT with cardiac defects was not completely assumed.

*Cardiac Axis: in the initial stages of pregnancy the cardiac axis assessment was regarded as a portion of fetal heart examination" the normal cardiac axis is defined at a 45-degree angle

to the left of the midline with a range of ±20 degrees "[4]. Abnormal cardiac axis associated with CHD have been seen in many studies [5, 6] Fig. (1).

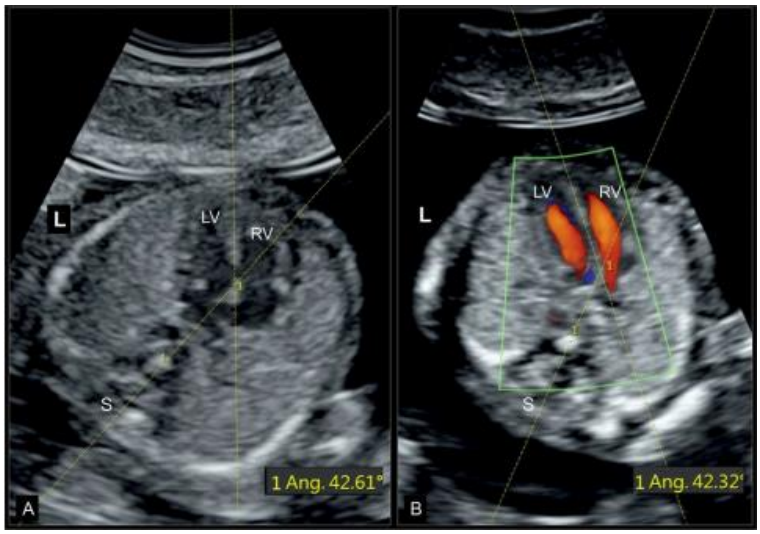


Figure 1:" fetus at 13 weeks' gestation with normal cardiac axis in B mode (A) and color Doppler (B).In a four-chamber view of the heart the cardiac axis by the angle of two lines; the first one begin at the spine (S) posteriorly and ends in midchest anteriorly, which divided the chest into two equal halves, the second one runs through the ventricular septum. This angle is 42 degrees (normal). RV, right ventricle; LV, left ventricle; L, left" [1].

*Reversed A-Wave in Ductus Venosus: Normal doppler morphology of ductus venosus (DV) is triphasic wave, the first peak represent systole (S); the second one represent diastole (D) and a nadir represent atrial contraction (A). Fig. (2)

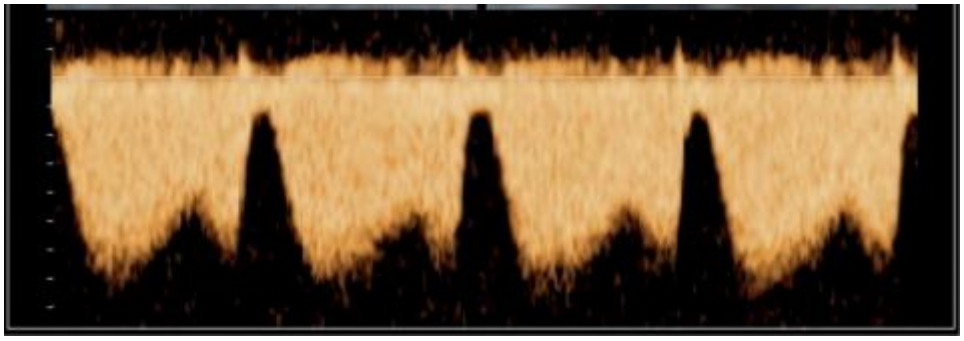


Figure 2: Normal doppler wave in ductus venosus[1].

"Matias *et al.* [7] were the first to observe the link between the occurrence of a reversed A-wave in the DV at 11 - 13 weeks and CHD in fetuses with a thickened NT" Fig. (3).

Common CHD with its finding on ultrasound:

Septal Defects

Ventricular Septal defects

Lonely seen ventricular Septal Defects (VSD) are the major occurrence heart malformation , " which account for approximately 30% of heart abnormalities seen in new

born, with an incidence of 1% to 2%" [9],although VSDs seen in isolation ,they may be seen associated with other cardiac malformation and other extracardiac anomalies .
Defects may be seen in each part of septum, with membranous / perimembranous occur more commonly .Defects that occur in muscular part of the septa may be seen as outlet ,inlet , apical or trabecula defects.

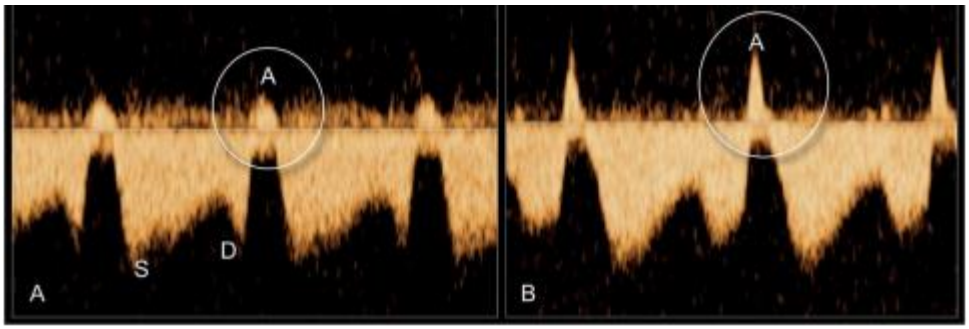


Figure 3: Pulsed doppler in two fetuses (A, B) at 13 week gestation with cardiac defect show reverse A wave. *Tricuspid Regurgitation (TR): may be seen in all gestational age and may be transient. TR seen at 11^{th} – 13^{th} week's gestation have an association with CHD [8].

The four-chamber view is employed for detection VSD in prenatal screening, we used subcostal four chamber view utilizing high frequency probe (5 -7.5MHZ) with beam vertical to the septum. The defect in septa seen as a dark, hypolucent region (Fig. 4). We must see a true VSD in further than one plane, so we move the transducer from anterior aspect to the posterior aspect of the heart [10,11]. There is a sign called" T sign "which help in distinguishing a true VSD from wasting signal. In a "T sign "the anechoic region is enclosed by an abruptly distinct area of the septa which appear hyperechoic.

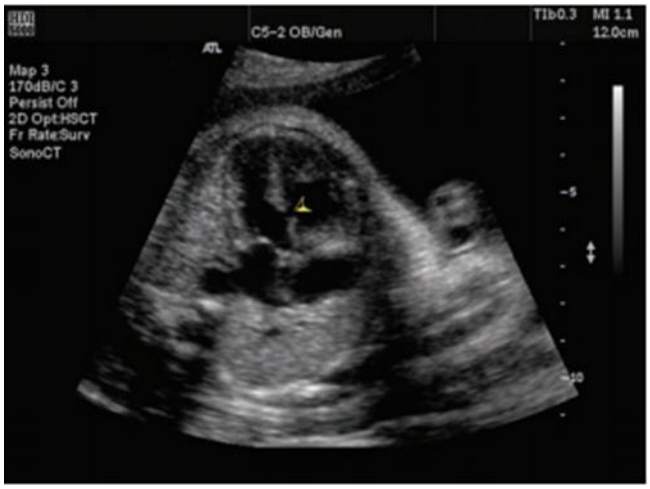


Figure (4): A four-chamber view showing a VSD (arrowhead) [12].

Although four chamber view is useful we need to sweep all septum which accomplished by utilizing long axis view of both outflow tract (left and right) & the short-axis view of foramen ovale and patent ductus arteriosus which causing equal pressure between both

ventricles .both ventricles , also we utilized both color and pulsed Doppler which may aid in diagnosis ,although it cannot see flow in all cases owing to the existence of foramen ovale and patent ductus arteriosus which causing equal pressure between both ventricles .

Atrial Septal Defects (ASD)

Regarded the 5th mostly seen congenital defect of cardiac defect about 7% of cardiac lesions [13]. ASD divided according to their location in septum either membranous or muscular with the "ostium secundum defects "are most frequent form, the" Ostium primum defects "are the 2nd frequent form and mainly seen as a part of multifarious abnormalities like atrioventricular defect. The ASDs which called Sinus venous ASDs include a defect which seen close to either the" superior vena cava "or "inferior vena cava". Using subcostal four chamber view to see ASD but sometime the diagnosis is misinterpreted by the existence of patent foramen ovale.

For diagnosis of ASD by ultrasound we demonstrate an anechoic zone in septum secundum that seen adjacent to the foramen ovale. To diagnose ostium primum ASD, the atrial septal part proximately over the atrioventricular valves is lacking and the septal leaflets of tricuspid valve appear to insert at same level of anterior mitral valve leaflet while in normal fetal heart insert apical to it. Also utilizing color doppler aid in diagnosis but is of limited use in diagnosis of small defects on account of the existence of turbulent normal flow seen across the foramen ovale.

Atrioventricular Septal Defects (AVSD)

Comprises a spectrum of heart malformation including grouping of abnormalities of the atrioventricular valves, ostium primum ASD and VSD. It divided into complete, intermediate or partial defects.

By using the four chamber view either subcostal or apical ,the majority of AVSDs possible to seen easily appearing as a big anechoic zone including the atrial septum (lower part) , the ventricular septum (upper part) , and a typical site of tricuspid and mitral valves, using color doppler can helping us to see the communication between four chambers.(Fig. 5)

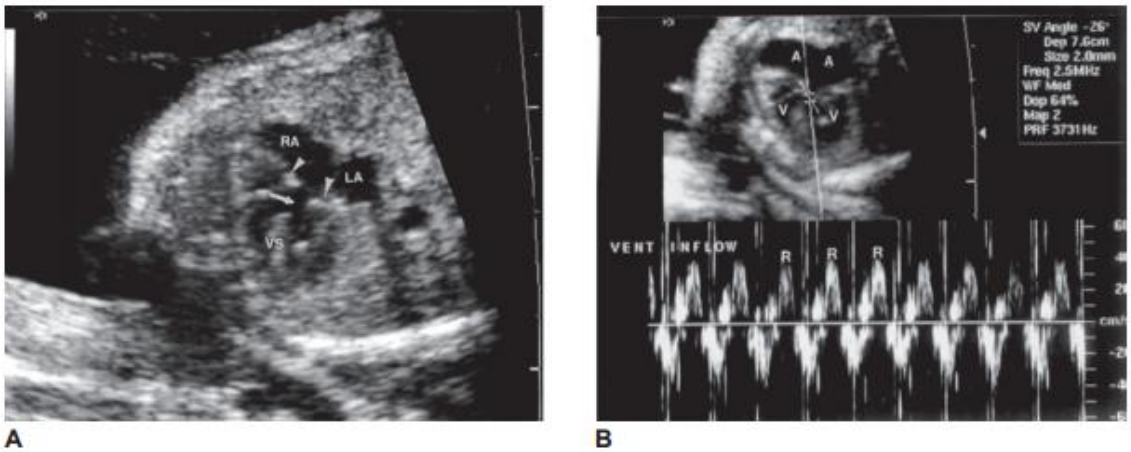


Figure (5): A: Four-chamber view showing endocardial cushion defect (arrow). RA, right arium; LA, left atrium; VS, ventricular septum. The common atrioventricular valve pointed by arrowheads. B: Doppler study shows valve incompetence. A, atria; V, ventricles; R, regurgitation jets [12].

Identification of intermediate and partial AVSDs are hard but demonstration of both VSD & ASD may give a clue to extra view of tricuspid and mitral valves. In AVSD we must look for aortic arch looking for anterior and superior displacement of arch which results in constricting of outflow tract of left ventricular. The deformity "goose-neck" is possible to demonstrate in viewing the left outflow tract in long axis [14].

Hypoplastic Left Heart (HLHS)

Is the greatest obstructive lesion of left side and common cause of death in a newborn? It consist of hypoplasia of left ventricle , aortic atresia, hypoplasia of ascending aorta , atresia and or hypoplasia of mitral valve, and a small size left atrium in some cases. It is accountable to about 7% of CHD [15]. As it is progressive condition sometime the sonographic finding may be late to the end of 2nd trimester.

On ultrasound examination the cardiac axis unchanged in HLHS. The subcostal four chamber view can demonstrate the big difference between sizes of two ventricle, we search for a band in enlarged or usual normal size RV to confirm LV hypoplasia. Owing to the hypoplasia of left ventricle, the apex of heart form by right ventricle (Fig. 6).

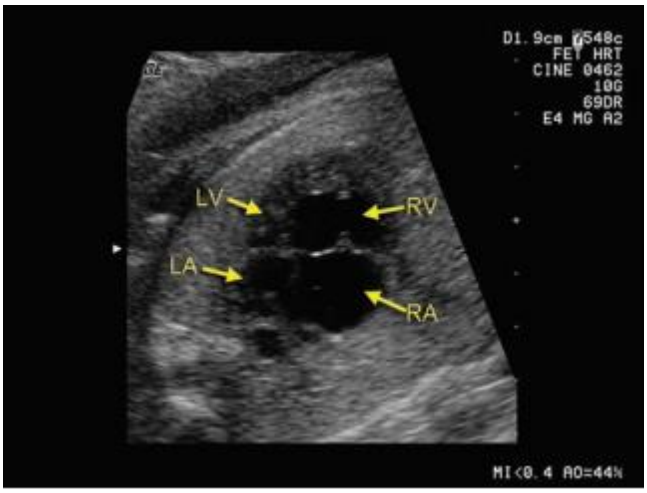


Figure (6) this four-chamber view showing enlargement of right atrium and ventricle. RA, right atrium; RV, right ventricle; LA, left atrium; LV, left ventricle [12].

Demonstrating right outflow tract by using short axis view to see the extent of aortic hypoplasia or aplasia, and additional three vessels view is used. Long axis view of left outflow tract or four chamber view use to see stenosis of mitral valve. Color and pulse doppler is used to demonstrate flow through aortic or mitral valves and also to assess flow through foramen ovale and ductus arteriosus as it is a ductal dependent condition.

Using M-mode as well to approve the diagnosis. For the diagnosis there are criteria which consist of" a left ventricular end-diastolic diameter of less than 9 mm with an aortic root diameter of less than 6 mm [16].

Right ventricular Hypoplasia (Pulmonary atresia)

Is infrequent condition, seen in about 0.1 - 0.4 in 10,000 new born baby. [9, 17].If it happened is a result of pulmonary atresia with a normal interventricular septum.

Demonstration of abnormally small right ventricle which seen by subcostal four chamber or apical view, assess the patency of tricuspid and pulmonary valves and may demonstrate

tricuspid regurgitation on doppler study. Right ventricular is ductal dependent so we must assess the patency of foramen ovale and truncus arteriosus.

Tricuspid Atresia

It has resulted in a hypoplasia of right ventricular owing to absence of flow from right atrium; it is a progressive condition which deteriorates as pregnancy proceeds.

By ultrasound it looks as a hyperechoic thickened valve, which once interviewed by Doppler study, displays absence of flow on each side of the valve. Ventricular septum should carefully asses looking for VSD, also pulmonary artery and valve should checking for presence of atresia.

Coarctation Of The Aorta

Is a constricting of aorta at level of aortic arch which results in outflow obstruction, according to its site as relative to ductus arteriosus it is divided to preductal, ductal or postductal with the ductal or postductal are more common type. Coarctation contribute to about 7% of CHD.[18].It is commonly accompanying with other anomalies either vascular like berry aneurysms or extracardiac anomalies.

It is one of difficult condition to diagnose prenatally. In second trimester screening, the visualization of right ventricular enlargement without other abnormalities should trigger the possibility of Coarctation of aorta. Hornberger et al. in 1994 found that the ratio of rt. ventricular size to that of lt. ventricle of 2.25 seen in fetuses who have coarctations as related to that ratio of 1.25 for healthy infants. Also, a ratio of diameter of the pulmonary artery to that of aorta was increases , with ratios approaching two in fetuses have coarctation of aorta and a ratio about 1.25. Follow the aorta for any narrowing and best seen in longitudnal view of the body when the arch and whole length of the aorta are seen well (Fig. 7). When we see the narrowing segment we measure postobstruction velocity by doppler study which appear higher than normal. At least the structure of aortic valve should be assessed with caution because of association of bicuspid valve with coarctation better seen by short axis right outflow tract.

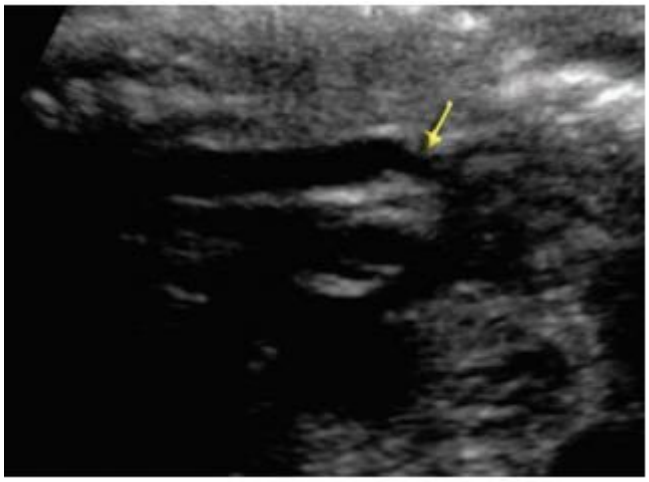


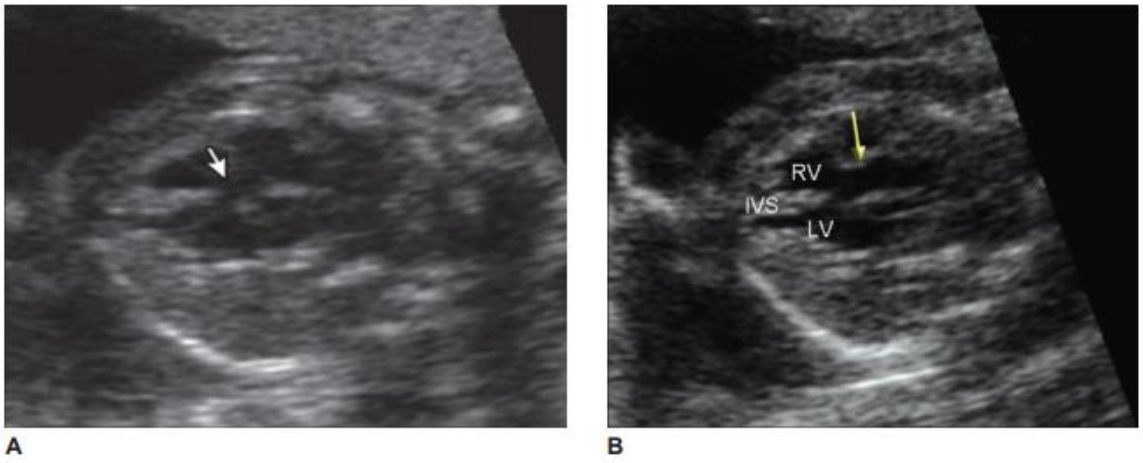
Figure 7: The postductal coarctation (arrow) of the aorta. [12].

Tetralogy Of Fallot (TOF)

Is one of commonest form of CHDs, seen in up to 7% of (CHD),it consist of four major component which are : * perimembranous VSD; * overriding aorta; * pulmonary stenosis or atresia; and *right ventricular hypertrophy. Ultrasonographic diagnosis of it started from demonstration of a VSD (perimembranous type) in both four chamber (subcostal and apical) view with using of color doppler which confirm the diagnosis. We make a cranial turning of the ultrasound probe to get a five-chamber view for demonstration of aortic root overriding the septal defect. Further assessment of VSD and overriding of aorta we need visualization of left ventricular outflow tract by long axis view. Overriding aorta that seen in tetralogy versus may be confused with a true double-outlet right ventricle (DORV); though, using "50%" rule may be useful. "If more than 50% of the aorta overrides the right ventricle, DORV is more likely [9].

Outflow tract of right ventricle need to be assessed ,in TOF we may see three kind of pulmonary artery abnormalities :pulmonary stenosis ; pulmonary atresia with major aortopulmonary collaterals and atresia of pulmonary artery with a patent ductus which can be demonstrated using right outflow tract short axis. If we see a small pulmonary artery as comparing to aortic root which support the diagnosis of TOF? The diameter of pulmonary artery is between 40% and 55% appear to be lesser than diameter of aorta in one series of fetuses with TOF examine in third-trimester; this variances showed to be more as pregnancy advanced (Fig. 8).

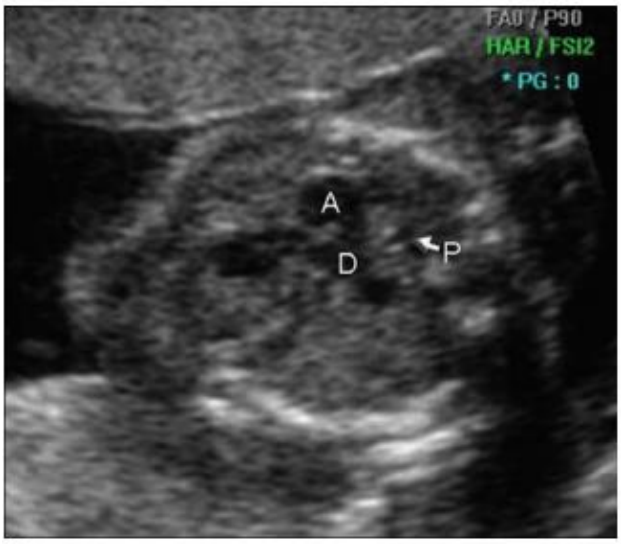
Using doppler study we see the velocity in pulmonary artery is higher comparing to the aorta. If atresia of pulmonary artery present rather than stenosis, a retrograde flow seen with flow absence through the valve itself, this flow is accompanying with increased turbulence. Right ventricular hypertrophy may not be seen prenatally.



A B

Ebstein's Anomaly

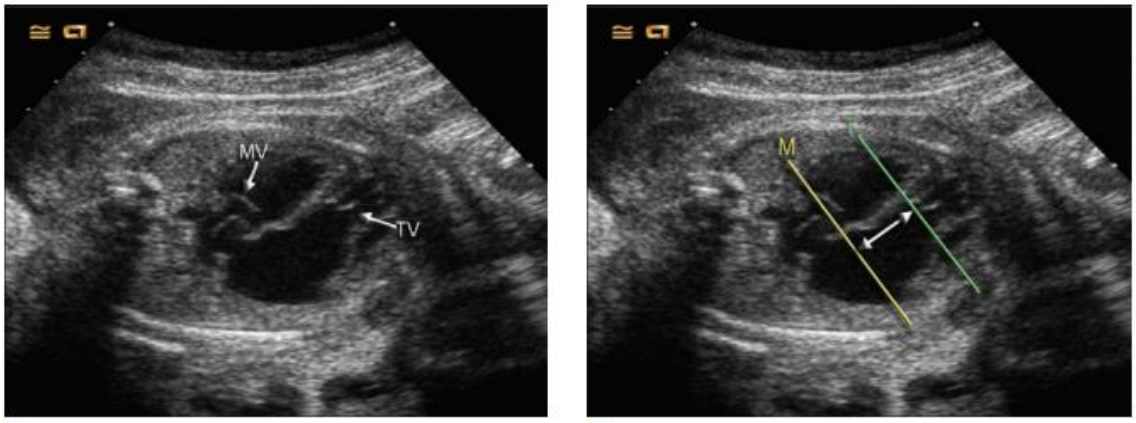
In this kind of anomalies the leaflets of tricuspid valve (septal and posterior) appear to be shifted apically from their usual normal place at AV junction into right ventricle causing enlarged right atrium and small right ventricle. So a complicating of this, the tricuspid valve aberrant leaflets appear adhesive to the wall of ventricle to variable extent, which result in a small movable part of the valve cusps. This in sequence, cause an important tricuspid regurgitation or, if the valves are immovable, a functional tricuspid stenosis seen.



C

Figure 8: Tetralogy of Fallot. A: This VSD (arrow) is part of the TOF. B: Long-axis view of the heart showing the aorta overriding the intraventricular septum and VSD. RV, right ventricle; LV, left ventricle; IVS, interventricular septum. C: Long-axis view of the heart showing a small pulmonary artery (P) .A,aorta; DV, ductus venosus [12].

Right atrial enlargement becomes more due to the presence of tricuspid stenosis or regurgitation [19]. Ebstein's anomaly is uncommon in which it often results in massive dysfunction in fetus, including cardiomegaly, hydrops, and arrhythmias [19]. The key finding in prenatal diagnosis is the presence of enlarged right atrial with apically shifted tricuspid valve with degree of a tricuspid regurgitation which measure with doppler study (Fig. 9).



A B

Figure (9): A: An apical four-chamber view showing mitral valve (M) and apically shifted tricuspid valve (T). B: Measuring the distance between the mitral and tricuspid valve indicates the severity of the inferior offset [12].

Because of this displacement there will be cardiac levorotation of the heart which produced by the enlarged right atrium, the leaflets of tricuspid valve are infrequently tethered which

seen on subcostal approach as either abnormal numbers of attachments or thickening of the chordate (Fig. 10).

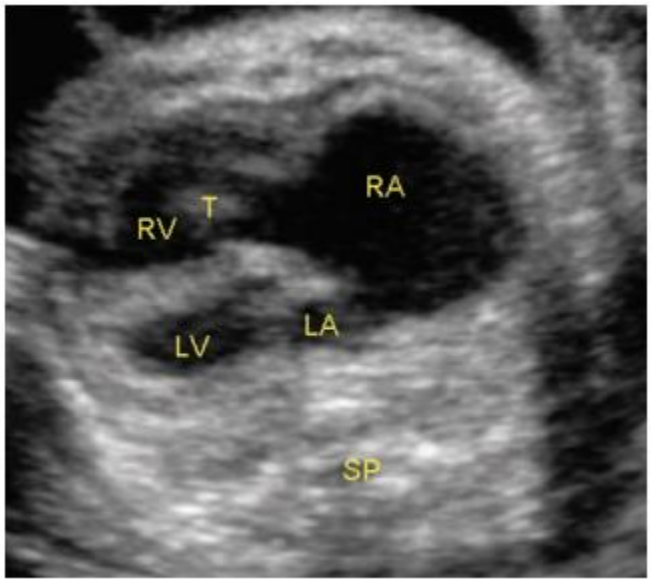


Figure 10: This four-chamber view subcostal approach showing levorotation, a thickened tricuspid valve (T), and an enlarged right atrium (RA) which causes left atrium (LA) compression. LV, left ventricle; RV, right ventricle; SP, spine [12].

Transposition of the Great Arteries

Grouped into 2 discrete forms: "complete transposition, or d-transposition, and congenitally corrected transposition or l-transposition". To represent the position of the aorta relative to the pulmonary artery we utilized the d- and l- , as d- means right and l- means left. In each type, the aorta arises from the right ventricle and the pulmonary artery arises from the left ventricle. The diagnosis of complete TGA is difficult prenatally, in complete transposition type, the four chamber subcostal and apical views appear normal, as the correlation between heart chambers is unchanged, so we depend on outflow tracts visualization. The pulmonary artery and central aorta seen either two circular structures adjacent to each other or as two vessels parallel to each other's (Fig.11) in a short-axis view of the right outflow tract, while normally the pulmonary artery demonstrating encircling the central aorta.

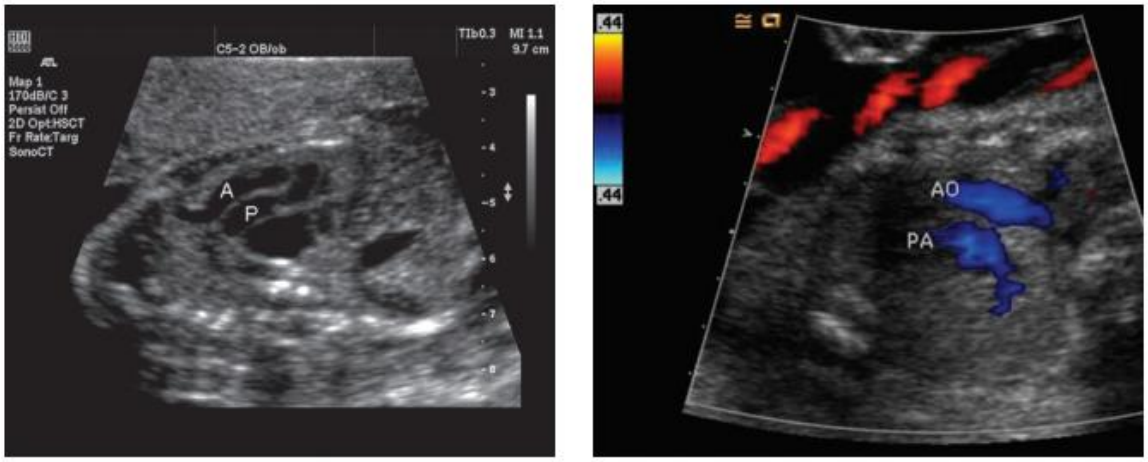


Figure 11: A: short axis view demonstrate the aorta (A) and pulmonary (P) arteries coming from the heart in a parallel course. B: Color Doppler imaging helps in demonstrating the vessels, confirming flow direction in the aorta (AO) and pulmonary artery (PA [12].

Regarding congenitally corrected transposition (CCTGA or l- transposition) in general is uncommon condition. In this condition, there are disagreeing in the connection of both atrioventricular and ventriculoarterial chamber. So the diagnosis depends on identification of chamber, the moderator band and tricuspid valve which displaced apically is seen attached to the left rather than the right atrium. The pulmonary and aorta arteries seen in a parallel arrangement.

Truncus Arteriosus

Is a part of uncommon conotruncal defects which occur in about 1% of fetal heart condition [20]? The truncoconal ridges which in normal situation seen split the truncus arteriosus into aortic and pulmonary trunks in this condition be unsuccessful to fuse so result in one great vessel arise from above the interventricular septum. At the upper portion of interventricular septa a VSD is present below the location of the truncus.

The diagnosis of it depend on visualization of single vessel overriding a VSD which is demonstrating in five-chamber view , as well as the demonstration of VSD in apical or subcostal view ,also the left ventricular outflow tract long axis view can be helpful in demonstration of a VSD with a single vessel overriding it. Lastly, the course and patency of whole length of the aorta must be seen well , up to 30% have a right-sided aortic arch , and possibility of an interrupted aortic arch also occur [9].

Double-outlet Right Ventricale (DORV)

It covers a cluster of conditions with greater than 50% of both the aortic root and pulmonary artery rise from morphologically apparent right ventricle, frequently above a VSD perimembranous type.(Fig .12)

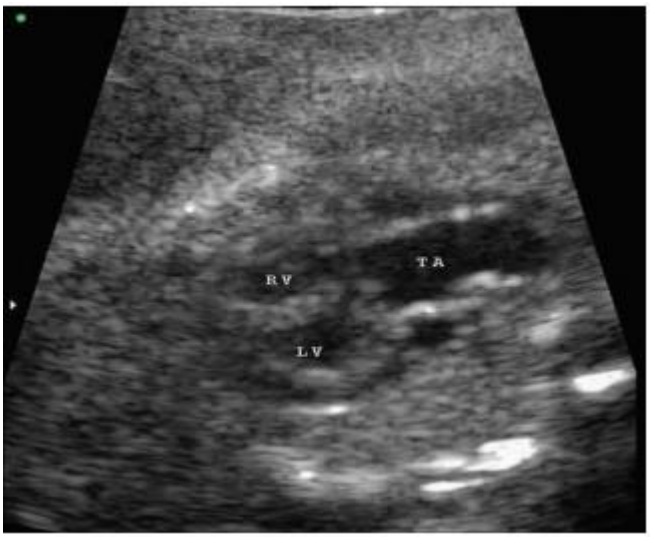


Figure 12: Truncus arteriosus (TA). An extended four-chamber view demonstrating the common truncus great vessel, arising from both the right ventricle (RV) and left ventricle (LV) [12].

About four main types resent: "* VSD-type, there is DORV with a subaortic VSD. * Fallot-type, DORV with subaortic or double committed VSD and pulmonary stenosis.* TGA type, in which there is a DORV with a subpulmonary VSD. * Noncommittal VSD-type, in which the VSD is remote from either great vessel [20].

Diagnosis by ultrasound can be difficult and the difficulty is seen in differentiate between it and conditions like tetralogy (particularly the Fallot-type) or HLHS. The long axis view of the pulmonary and aorta artery, a VSD all of them may be seen in this view. The arrangement of aorta and pulmonary artery side by side which appear as parallel outflow tracts which are similar to that visualized in TGA. Pulsed Doppler study is utilized to rule out hypoplasia and coarctation of both the pulmonary artery and the aorta.

Conclusion

Screening ultrasound examination of the fetal heart play an important in diagnosis of CHD. Early assessment of the fetal heart as a screening process must be performed in pregnant woman who have a history of CHD specially in her family ; any other fetal noncardiac anomaly, increased nuchal translucency, tricuspid regurgitation, reversed atrial waveform in the ductus venosus, abnormal cardiac axis ; and hydrops. A good training and experience of the operators are considered an essential factors to a successful demonstration of fetal cardiac defects.

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