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The place of ultrasonography in detecting fetal heart malformations

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Abstract

Most newborns with cardiac malformations are born without obvious risk factors, so the prenatal diagnosis depends on screening protocols. Fetal cardiac ultrasound should be a part of every ultrasound examination, especially when other malformations are discovered. Contrary to neural tube defects or trisomy 21, detecting fetal heart malformations implies therapeutically termination of pregnancy in only half of the cases. Echocardiographic exam (2D, 3D, Doppler) allows the diagnosis of 90% of cardiac defects, given performing machinery and an experimented examiner.

Keywords: echocardiography, congenital heart malformations, standard sections, prenatal screening

Rezumat

Majoritatea nou-născuţilor cu malformaţii cardiace se nasc din mame fără factori de risc evidenţi, aşadar depistarea acestora înainte de naştere este dependentă de protocoalele de screening. Ecocardiografia fetală ar trebui inclusă în fiecare examinare ecografică, mai ales în condiţiile depistării altor malformaţii. Spre deosebire de defectele de tub neural sau trisomia 21, depistarea unei malformaţii cardiace fetale nu implică decât în jumatate din cazuri întreruperea terapeutică a sarcinii. Examinarea ecocardiografică (2D, 3D, Doppler) permite depistarea unui procent de 90% dintre malformaţiile cardiace, în condiţiile unei aparaturi performante şi a unui ecografist experimentat.

Cuvinte cheie: ecocardigrafie, malformaţii cardiace congenitale, secţiuni standard, screening prenatal

Introduction

The development of the fetal heart bears the influence of genetic and environmental factors, maternal diseases and infections. Approximately 70-80% of congenital heart defects have multiple determinants [1,2]. Congenital cardiac anomalies are frequent, with an incidence of 8/1000 births and they are associated with an increased morbidity and mortality rate in fetuses and newborns [3,4,5]. Congenital heart defects are in 48% of cases identified with chromosomal anomalies [6].

Examining the fetal heart implies access to high resolution machinery, using the following examination modes: bidimensional, Doppler, 3D, 4D and an experienced ultrasonographist. Fetal echocardiography is recommended beginning with the 18th week of gestation, major cardiac anomalies being detectable from the 12-13 week of gestation. Each extra week of gestation allows a better visualization of the fetal heart.

The therapeutic decision to be taken in case of detection of a congenital heart anomaly depends on its association with other anomalies. Malformative syndromes frequently require therapeutic termination of pregnancy, whereas an isolate cardiac defect implies informing the parents, the decision involving them directly. At present, over 90% of heart defects are not diagnosed because of the limited experience of sonographists, but when the examination is performed by a specialist, the fetal echocardiographist, the detection of heart defects exceeds 90% [7,8].

Using the traditional 4 chamber image of the heart may detect between 5 and 50% of cardiac malforma-

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tions [9]. Measurement of the long and short axis of the fetal heart increases the detection rate to 30% and the analysis of the ejection pathways raises the percentage of detection to 70-80%. Adding the colour and power leads to an increase to 90% of the rate of [10,11, 12].

Indications

The risk factors associated with congenital cardiac malformations can be fetal, maternal or family related. Amongst the fetal factors, the most important are extracardiac anomalies, chromosomal anomalies, cardiac arrythmias (atrioventricular block, premature beats), non-immune fetal hydrops, intrauterine growth restriction or polihydramnios. Regarding the maternal/family risks factors we mention: congenital cardiac anomalies in brothers/parents, exposure to teratogens (alcohol, lithium, amphetamines), maternal diseases (diabetes mellitus, collagenosis, phenylketonuria, maternal infections, family syndromes) [13,14]. Although the risk of congenital heart malformation is of 8/1000 births, it increases to 12/1000 births if either of the parents has such an anomaly [15]. Detection of a fetal anomaly implies a minute examination of the fetal heart given the increased risk of associating a cardiac defect.

Normal anatomy and fetal heart malformations

Examination of the fetal heart implies, firstly, determining the fetal position, then establishing the position of the fetal heart, verifying the atrio-ventricular connection, the atrio-ventricular valves, detecting anomalies. Evaluation of the fetal heart may be reduced to three main cardiac segments: the viscero-atrial site, the atrio-ventricular site, and the arterial trunk [16,17].

There are three types of viscero-atrial site anomalies:

1. *situs solitus* – it depicts the normal situation: the stomach to the left side, the right atrium to the right, the left atrium to the left
2. *situs inversus* – the stomach to the left, but the right atrium to the left and the left atrium to the right
3. *situs ambiguus* – the liver is situated at the median line, the stomach to the right or to the left. It comprises two syndromes: asplenia and polisplenia.

Four standard sections of the fetal heart are used for the evaluation of the atrio-ventricular site and the arterial trunk:

1. the four chambers image;
2. the ejection pathway of the left ventricle;
3. the three vessels image;
4. the aortic arch (fig 1).



Fig 1. Aorta – a longitudinal section: the ascending segment, the arch (with the neck vessells poited by arrowheads), the descending segment.

The 4 chambers image is the most important for the examination of the fetal heart and its proper evaluation is essential. It is modified in 60% of major cardiac anomalies [15,18]. It must be systematically analyzed where its dimensions, position, structure and cardiac function are concerned.

Dimension: the heart normally occupies 1/3rd of the thorax and it can be visually appreciated in most cases. A cardio-pulmonary index can be calculated (VN 0.5-0.6). A small index may be present in intrathoracic compressions (pleural collections, diaphragmatic hernias, thoracic masses), and an elevated index is present in cardiomegalies (fetal anemia, atrio-venous malformations, complete cardiac block, a decrease of cardiac contractility), dilation of one of the heart chambers or, a small thorax (various skeletal dysplasia) [14,19].

Position: the heart is, in most cases, placed in the left side of the thorax; the left atrium is the most posterior situated chamber, immediately anterior to the descending aorta, which is situated anterior to the thoracic backbone. The right ventricle is located behind the sternum. The interventricular septum makes a 45 degree angle with the median line of the thorax, with the apex towards the left (fig 2).

There are two types of position anomalies:

a. a different location inside the thorax: in the right thorax (shift to the right, dextrocardia) or on the median line;

b. a change of the cardiac axis, to the left (a right intrathoracic mass, cardiac defects, left pulmonary hypoplasia), or to the right (right pulmonary hypoplasia, left intrathoracic mass).



Fig 2. Four-chamber image of the heart: 2/3 of the heart appers in the left hemithorax, there are 2 similar atria and two similar ventriculi, intact intraventricular septum. the apex is 45 degrees to the left (levocardia).



Fig 3. The heart in four chambers. The valvular muscles appear at the site of the two ventricles (arrows).

Structure: two atria of approximately equal dimensions, 2 ventriculi of approximately equal dimensions, atrio-ventricular valves (fig 3). A moderator band is described towards the apex of the right ventricle – the trabecular muscles. Inside the pericardium usually there is a thin layer of liquid. "The crus" (the heart cross) is formed of the atrio-ventricular septum and the septal leaflets of the mitral and tricuspide valves. The tricuspide valve is placed more apically than the mitral. The intraventricular septum is intact from the apex up to the crus. The interatrial septum is attached to the crus and it presents in the middle third with a defect – the foramen ovale. The pulmonary vessels shed in the left atrium.

Ventricular anomalies are classified as follows:

- unequal ventricular dimensions: the RV (right ventricle) bigger than the LV (left ventricle) in a normal term oregnancy, aortic coarctation, tricuspidal insufficiency, mitral stenosis; RV smaller than LV in mitral insufficiency, severe aortic insufficiency, tricuspidal stenosis);
- unique ventricle.

Crus anomalies are: the unique atrioventricular valve (mitral or tricuspidal atresia, atrio-ventricular septum defects).

Other structural anomalies are: interventricular septum defects (membranous or muscular), interatrial septum defects, cardiac tumors, venous return flow, left atrium isomerism.

Function: The atria and ventriculi contract synchronously and regularly with a frequency of 120-180 beats/minute. During the Doppler examination there is an equal ventricular filling, without any significant atrioventricular regurgitation.

Examination of the arterial trunk (great vessels) allows the detection of 40% of cardiac malformations, amongst which the tetralogy of Fallot, the great vessels transposition and also the presence of a common arterial trunk. The great vessels must be examined where their dimension, position, structure and function are concerned. The sections used for such examination are the following:

a. standard: the ejection pathway of the LV (the origin of the aorta) (fig 4), the three vessel plane (fig 5), the aortic arch;

b. other: the long axis of the LV, the long axis of the aortic arch, the short axis of the LV, the aortic arch and ductus venosus images, the bicave plane and the aorto-tricuspidal plane.

Dimension: normally, the pulmonary artery (fig 6) has a diameter slightly greater than the aorta. The ratio between the pulmonary artery dimension and the origin of the aorta is of 1–1.09 throughout the whole pregnancy [20].



Fig 4. The ejection pathway of the left ventricle.



Fig 5. The three vessel plane.



Fig 6. The ejection pathway of the right ventricle – origin of the pulmonary artery.

Dimension anomalies are: a larger PA (pulmonary artery) – absence of the pulmonary valve, PA narrower than the aorta (pulmonary stenosis, tricuspidal flow reduction – tricuspidal stenosis or atresia, Ebstein disease), the aorta much narrower that the PA: aortic stenosis, aortic coarctation, aortic interruption.

Position: normally, the pulmonary artery is located to the left, anterior and cranial to the origin of the aorta. Position anomalies are:

- aortic valve to the right, anterior and superior to the pulmonary artery (double exit RV, great vessels transposition);
- aortic valve to the left, anterior and superior to the pulmonary artery (corrected great vessels transposition);
- parallel great vessels (double exit RV, great vessels transposition, corrected great vessels transposition)

Structure and function: normally, the valves open freely, there is blood flow through all the valves at a speed of under 100 cm/sec; there is no regurgitation. The encountered anomalies are: the opening restriction of the pulmonary/aortic valve with an increased flow (pulmonary or aortic stenosis), pulmonary regurgitation (pulmonary valve absence), aortic regurgitation (RV-aorta tunnel).

The examination of the great vessels must provide the answer to three questions:

1. Is there a normal crossing of the aorta and the pulmonary artery? Normally, the aorta has a trajectory cranial and to the right and the pulmonary artery has a trajectory to the posterior, thus the two vessels cross each other in an angle which is almost right. The ramification of the pulmonary artery occurs immediately after having left the RV, serving for its differentiation from the aorta. If this crossing does not exist and the two arteries are parallel to one another, this is a sign of double exit RV or great vessels transposition.

2. Are there discrepancies between the dimensions of the two vessels? Evaluation of the dimension of the two vessels serves for the diagnosis of anomalies such as Fallot's tetralogy, where the aorta is larger than the pulmonary artery which is atresic. In the case of the common arterial trunk, only one arterial canal can be visualized and not two separate vessels. Fallot's tetralogy, the common arterial canal, the LV hypoplasia with transposition determine an enlargement of the aorta, whereas the LV hypoplasia and aortic coarctation determine a decrease in dimensions.

3. Are the great vessels in a normal relationship with the interventricular septum? Normally, the aorta leaves the LV having a trajectory to the right; the anterior wall is in connection with the interventricular septum and the posterior wall with the anterior leaflet of the mitral valve. The crossing of the interventricular septum by the aorta occurs most frequently in Fallot's tetralogy, pulmonary atresia with a ventricular septum defect, arterial trunk and double exit RV.

Doppler Ultrasound

Used for examining the fetal heart, Doppler ultrasound has several purposes: determining the direction of the blood flow, identifying the vessels, the left-right shunts and the hypoplasic heart. In the case of left heart hypoplasia, Doppler exam reveals the absence of flow in the LV and reverse flow in the aortic arch. In some situations, such as aortic coarctation, the tetralogy of Fallot, color Doppler may assist in confirming the diagnosis. Color flow Doppler proves its value in examining obese patients, where acquiring a four chamber image or detecting the intraventricular septum may be difficult, especially in membranous locations, detecting an atrioventricular canal, tricuspidal or mitral regurgitation.

Pulsed wave Doppler is useful for evaluating the valvular flow, especially in the case of valvular stenosis. Doppler evaluation of the aortic and pulmonary valves reveals a single apex, and for the mitral and tricuspide - two apexes. In valvular stenosis an increase in the peak flow occurs [21].

Conclusions

Fetal echocardiography is a quick, inexpensive and non-invasive technique for detecting fetal cardiac malformations. It assumes the use of high performance machinery and an experimented ultrasonographist. Heart examination is a must, especially when malformative pathology has been diagnosed. The therapeutic decision in case of heart malformation detection depends on its association with other malformations and belongs, eventually, to the parents. Evaluation of the fetal heart implies visualization of multiple incidences in 2D, 3D, Doppler modes, the classical four chambers image being insufficient nowadays.

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