Early detection of fetal cardiac anomalies by transvaginal echocardiography

Bambang Yudomustopo

Department of Obstetrics and Gynecology Faculty of Medicine University of Trisakti
Department of Obstetric and Gynecology Pertamina Central Hospital

ABSTRACT

The fetal cardiac anomalies (FCA) is mentioned as congenital heart disease (CHD) or congenital heart malformation (CHM). The incidence of the FCA is around 8 infants per 1,000 birth. CHM are responsible for most infant dead in the first year of life. It was not until the late 1970s that refinements was done on the image resolution of fetal echocardiography. The detection of fetal cardiac abnormalities in high risk groups and the attempt to scan the whole pregnant population aroused great interest. The most important disadvantage and therefore the major limitations of first trimester echocardiography is the development of CHD later in intrauterine life. With the introduction of high frequency transvaginal transducer, it become possible to visualize the first trimester fetus in detail. Furthermore, sonographic examination at this stage of pregnancy should be complemented by tests such as karyotyping and DNA testing. It seems clear that the capacity of high frequency transvaginal probes is sufficient for performing echocardiography examination in the first trimester, to do early diagnosis of fetal cardiac abnormalities.

Keywords: Fetal cardiac, anomalies, transvaginal echocardiography, early detection, first trimester

Transvaginal ekokardiografi untuk deteksi dini anomali jantung janin

ABSTRAK

Anomali jantung janin (AJJ) dapat berupa penyakit jantung bawaan (PJB) atau malformasi jantung bawaan (FJB). Insidens dari AJJ sekitar 8 per 1,000 kelahiran. PJB merupakan salah satu penyebab kematian bayi. Sejak akhir tahun 1970 mulai dapat digambarkan jantung janin dengan ultrasonografi. Sejak masa itu, deteksi kelainan jantung janin mulai giat dikerjakan. Kekurangan terbesar dari pemeriksaan ekokardiografi pada trimester pertama kehamilan adalah terjadinya PJB pada masa kehidupan intrauteris. Dengan demikian memungkinkan pemeriksaan dan pemindaian (scanning) pada wanita hamil berisiko tinggi. Dengan ditemukan teknologi transjuser transvaginal yang berfrekuensi tinggi, memungkinkan pemeriksaan kelainan jantung janin pada trimester pertama. Selanjutnya pemeriksaan sonografi ini harus dilanjutkan dengan pemeriksaan DNA. Pemeriksaan transvaginal fetal ekokardiografi sangat bermanfaat untuk deteksi dini AJJ.

Kata kunci: Jantung janin, anomali, transvaginal ekokardiografi, deteksi dini, trimester pertama
INTRODUCTION

The fetal cardiac anomalies (FCA) is mentioned as congenital heart disease (CHD) or congenital heart malformation (CHM). The incidence of the FCA is around 8 infants per 1000 birth.(1) CHM are responsible for most infant dead in the first year of life. It was not until the late 1970s that refinements was done on the image resolution of fetal echocardiography. This method allows to evaluate more detailed of fetal cardiac anatomic structures and related malformations. Visualization of normal heart anatomic structures and subsequently the recognition of fetal heart malformations improved rapidly from this time onward.(1) The detection of fetal cardiac abnormalities in high risk groups and the attempt to scan the whole pregnant population aroused great interest.(2) Most centers performed fetal echocardiography at around 16-20 weeks gestation and used B-mode or M-mode transabdominal transducers. With the introduction of high frequency transvaginal transducers, it became possible to visualize the first trimester fetus in detail. The use of first trimester transvaginal echocardiography to detect or to make the early diagnosis of fetal cardiac anomalies has become more popular.

Despite several studies that stated the fetal heart examination performed in the first trimester, its use currently is still limited to a few specialized centers. (2) First trimester fetal echocardiography is defined as an attempt to visualize fetal heart anatomic structures before 14 weeks of gestation. The aim of this article is to describe the development of transvaginal fetal echocardiography, and doing early diagnosis of fetal cardiac anomalies. As well as the possibilities and limitations of such modalities, and finally to defined the indications in which the first trimester fetal echocardiography could be done.

Visualization of fetal heart before 10 weeks of gestation

Organogenesis takes place in the first 8 weeks of human life. In this relatively short period, which corresponds to 10 completed weeks of gestation, all of the major organs systems are developed. At the end of this period, most organs have reached a size at which they can be visualized by sonography. The cardiovascular system begins to develop within the lateral intra embryonic mesoderm at 4 weeks gestation age (GA). (3) Then the primitive heart tube is formed and begins to beat shortly after 5 weeks completed gestation. This primitive myocardial heart tube undergoes a process of looping, remodeling, and septation that transforms its single lumen into 4 chambers of the definitive heart. The final process of closure of the ventricular septum and ultimate formation of the atrioventricular valves is finished at 10 completed weeks of gestation. Sonographic studies of the fetal heart, before GA 10 completed weeks, have to take into account that morphologic structures are still unfinished. Examinations before this GA are, therefore, studies of the developing embryo rather than diagnostic tools.

The critical period of heart development is from 20th day after fertilization. Numerous critical events occur during cardiac development, and deviation from the normal pattern at any time may produce one or more congenital heart anomalies. Partitioning of the primitive heart results from complex results defects of cardiac septa which are relatively common, particularly VSD. Some congenital anomalies result from abnormal transformation of the aortic arches into the adult arterial pattern. (4,5)

First trimester diagnosis of fetal cardiac anomalies

The first published studies on the diagnosis of the heart malformations in first trimester fetus were case reports. The types of malformations diagnosed were mainly defects affecting the 4 chamber view, such as large ventricular septal defects (VSD), atrioventricular septal defects (AVSDs). Other types of anomalies are asymmetry of the ventricles. Diagnoses of malformations solely affecting the outflow tracts before 14 weeks of gestation are scarce, suggesting that these types of defects are more difficult to diagnose in the first trimester.

In two reports concerning malformations of the outflow tracts were, dextrocardiac or a large VSD. Except two reports, all authors described fetal heart defects in first trimester fetuses.
As shown in Table 1, using transvaginal probes, suspected heart malformations were due to increase nuchal translucency (NT), fetal cardiac arrhythmia, or family history of cardiac malformations.\(^{(6,7)}\) Gembuch et al showed that in 48.6% of the fetuses in whom cardiac defects were shown on sonography in the first and early second trimesters, a chromosomal abnormality was present. In their view, karyotyping should be offered to all patients whom fetal cardiac malformations are suspected in early pregnancy.\(^{(8)}\)

It is clear, therefore that there are many advantages and disadvantages of the first trimester diagnosis of fetal abnormalities. What is also clear, however, is the need for good quality equipment with a high frequency transvaginal transducer, experience in the technique of transvaginal sonography, and thorough knowledge of the embryological development of the first trimester fetus.\(^{(9)}\)

### Technical limitations of fetal cardiac evaluation during the first trimester

The most important disadvantage and therefore the major limitations of first trimester echocardiography is the development of CHD later in intrauterine life. Several malformations, such as pulmonary and aortic stenosis or coarctation and even hypoplastic left heart syndrome, can develop in second and even third trimester fetuses. Furthermore, some heart lesions such as cardiac rhabdomyomas or cardiomyopathy may not be present in the first and second trimesters and can evolve in later gestation or even after birth.\(^{(2,10)}\)

Bronshtein and Zimmer studied 36,323 transvaginal sonographic examination in early pregnancy. More than 99% of cases were evaluated at 14-16 weeks gestation. It was 173 cardiac anomalies detected, giving an overall incidence of 1 in 210 pregnancies. Ten fetuses had a cardiac anomaly which differed from the anomaly suggested by sonography. The sonographic diagnosis was confirmed after delivery or at postmortem in 90 cases. In the remaining cases, a postmortem examination was not possible because termination of pregnancy was performed by dilatation and curettage.\(^{(11)}\) Therefore it is important to inform to parents that some minor lesions can be missed in early pregnancy, even after detailed examinations. However rarely, a serious defect can develop after the second trimester. Thus after the first trimester echocardiography, a follow up examination in the second trimester should be performed (Figure 1).\(^{(12)}\)
Another disadvantage of the first trimester echocardiography is the possibly detection of defects that can resolve spontaneously in later pregnancy, such as muscular VSD. This can result in unnecessary anxiety in the parents. The knowledge that VSD usually remain undetected in the second trimester echocardiography probably because the sizes of the current available sonographic devices make it negligible on the first trimester examination. Because of the increased axial (0.3 - 0.4 mm) and lateral resolution of the modern broadband probes used in most studies, the fetal heart and its detailed structures up 1 mm can be readily visualized. Furthermore, a transvaginal probe only allows manipulation in one direction. If the fetal position is consistently unfavorable, the technique does not provide sufficient images of the fetal heart. This is the major reason for unsuccessful examinations. Finally, probably the most important reason that first trimester echocardiography is still rarely applied method is the need for highly skilled staff who have several years of experience in second trimester echocardiography and using high frequency transvaginal probes.  

Visualization rates of the full cardiac anatomic structures were 47.5% in an unselected population and 76.9% in a high risk population in study by Rustico et al, who examined fetuses between 13 and 15 weeks GA by transvaginal sonography. In that study, the high risk population was examined by operators with several years of experience, in contrast to the unselected population, which was examined by operators with less experience. The sensitivity for major heart defects was only 33%. Bronstein et al all found similar results, they reported 80% complete cardiac visualization on the initial scans, with a sensitivity of 55% for heart defects. Hugon et al, studied fetal cardiac abnormalities identified prior to 14 weeks gestation, at high population risk as a result of screening by nuchal translucency. They found the karyotype to be abnormal in 70/286 (24.5%) fetuses with normal fetal echocardiograms, and in 94/129 (72.9%) with abnormal or suspicious cardiac findings.

The last new techniques such as 3D, 4D sonography and the tools 3D multiplanar and 4D Spatio Temporal Image Correlation (STIC), should provide more information to do early diagnosis of fetal cardiac anomalies.

**Indications for Echocardiography in Early Pregnancy**

The importance of the mentioned limitations of first trimester echocardiography justifies restriction of its use to fetuses at high risk of having
cardiac abnormalities. Risk factors are generally accepted from the family members with congenital heart defects, increased risk of genetic syndromes in which cardiac malformations are present, increased nuchal translucency, fetal anomalies associated with cardiac defect, maternal diabetes, and the maternal use of cardiac teratogenic medications.\(^\text{(2,3)}\)

**CONCLUSION**

The cited reports on first trimester and early second trimester echocardiography shown at this early stage of pregnancy has great potential. Although the advantages of trimester transvaginal echocardiography seems of considerable value for couples at risk for having offspring with cardiac defect, this method is still limited in few specialized centers. Furthermore, sonographic examination at this stage of pregnancy should be complemented by tests such as karyotyping and DNA testing.

It seems clear that the capacity of high frequency transvaginal probes is sufficient for performing echocardiography examination in the first trimester, to do early diagnosis of fetal cardiac abnormalities. The capability of detecting and correctly diagnosing cardiac anomalies is highly dependant on the operator. It is hoped that recent advanced tools like 3D, 4D, multiplanar 3D image and 4D STIC can provide more information in the early diagnosis of fetal cardiac malformations.

**References**