Antenatal Diagnosis of Heart Diseases

- clinical experience and result analysis -

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Study realized in the research project MAMI no 41-042/2007, financed by the Roumanian Ministry of Education, Research and Youth

Abstract

Introduction. *Congenital heart disease have an incidence of 8/1000 live births, representing the most frequent congenital defects. Echocardiography is the only diagnostic tool for heart disease in fetus.* ***Material and method.*** *Between January 2009 and September 2010, 45 pregnant women with gestational age between 20 and 37 weeks were evaluated by echocardiography for fetal cardiac disease.* ***Result.*** *In 10 cases there was a normal pregnancy, in 31 cases a congenital heart disease was found and in 4 patients arrhythmia was diagnosed, among them in 2 cases cardiac secondary modification due to arrhythmia was found.* ***Conclusion.*** *Fetal echocardiography has a key role in antenatal diagnosis of heart disease, allowing the elaboration of the most efficient therapeutic strategy.*
Keywords: *fetal echocardiography, congenital heart disease, pregnancy*

Introduction

Congenital heart malformations are present in 0.8% of newborns, therefore they represent the most frequent congenital defects, having an important impact in neonatal mortality and morbidity[4,6,8,16]. Echocardiography is the only diagnostic modality of heart diseases in the fetus[25]. Progresses achieved in the domain of echography allow early determination of heart malformation diagnosis, as early as the first 20 weeks of intrauterine life[2]. The knowledge of diagnosis in the antenatal period allows the determination of an optimum postnatal therapeutic plan, and in certain cases offers to parents the possibility of interrupting the pregnancy[1,5]. Also, the discovery of cardiac defects should orient for the evaluation of the fetal karyotype[17].

Indications for fetal echocardiography can be classified in three major categories: fetal (extra-cardiac anomalies, genetic syndromes, hydrops, arrhythmia), maternal (mother with heart defect, teratogenic exposure, diabetes) and with family risk (child or parent with heart defect, Noonan syndrome)[11,17,19]. Most newborns with congenital heart malformations are born by mothers with low risk[8]. Therefore, the antenatal diagnosis of congenital heart diseases depends mostly on the obstetric ultrasound screening routine[15].

Prenatal diagnosis should be accurate so as to evaluate the therapeutic strategies and to properly inform the future parents. This involves a multidisciplinary collaboration between the gynecologist, pediatric cardiologist, neonatologist, cardiovascular surgeon[11].

Material and method

In the period January 2009 and September 2010 antenatal echo-cardiographies were carried out on a number of 45 patients, with the gestational age between 20 and 37 weeks. Patients were included in this study on the basis of the following criterias: modifications at the routine echocardiographic examination performed by the obstetrician, previous affected child or fetus, one of the parents with heart malformation, modifications of cardiac rhythm. The scanning of the fetal heart was carried out following this protocol[2,3,17,19,21]:

1. The initial step in any examination of the fetal heart is to establish laterality:
- heart on the left;
- apex to the left;
- stomach on the left.

2. Cardiac axis should be approximately 45º towards the left.

3. Chamber and vessel identity must be proved by defining their characteristic anatomic features.

4. The normal heart rate is between 120-160/min.

5. Other useful observation include:
- the most posterior chamber of the heart is the left atrium;
- the descending aorta lies between the spine and left atrium;
- the right ventricle can be slightly bigger than the left, especially in the 3rd trimester.

6. Offsetting refers to the differential insertion of the 2 atrioventricular valves:
- the septal leaflet of the tricuspid inserts slightly lower in the ventricle;
- both valves should be seen to open simultaneously during diastole in each cardiac cycle and close in systole;
- only the right ventricle has moderator band;
- the flap valve of the foramen ovale can be seen opening into the left atrium;
- a normal 4-chamber view will exclude most uncorrectable cardiac lesions and up to 1/3 of significant cardiac anomalies.

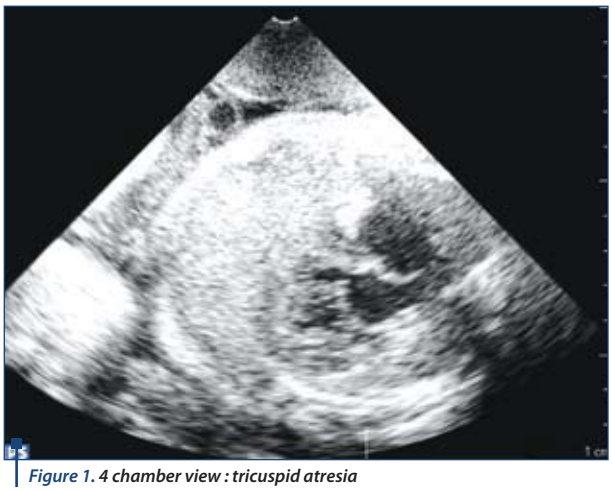

Figure 1. 4 chamber view : tricuspid atresia

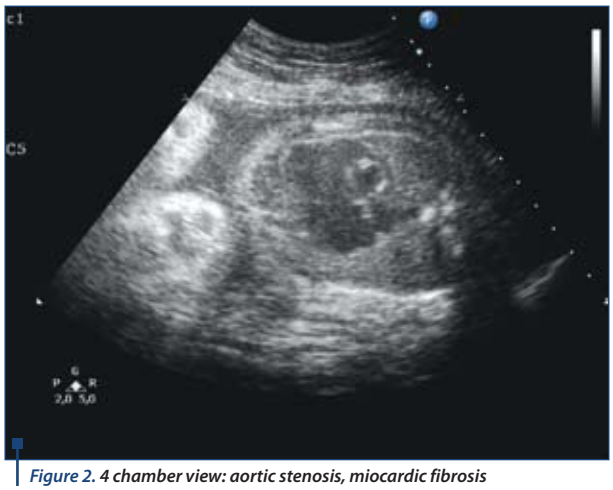

Figure 2. 4 chamber view: aortic stenosis, miocardic fibrosis

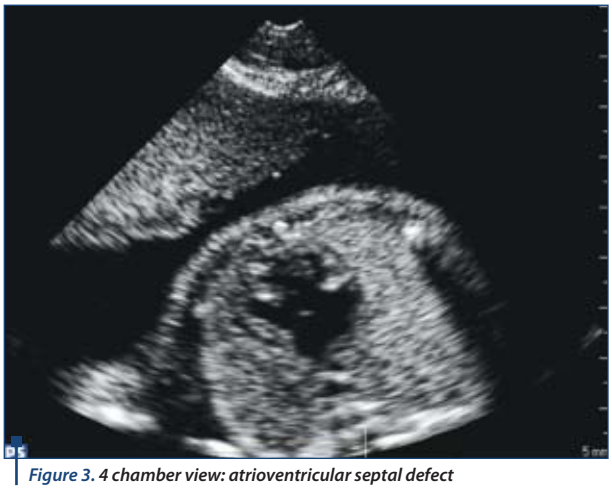

Figure 3. 4 chamber view: atrioventricular septal defect

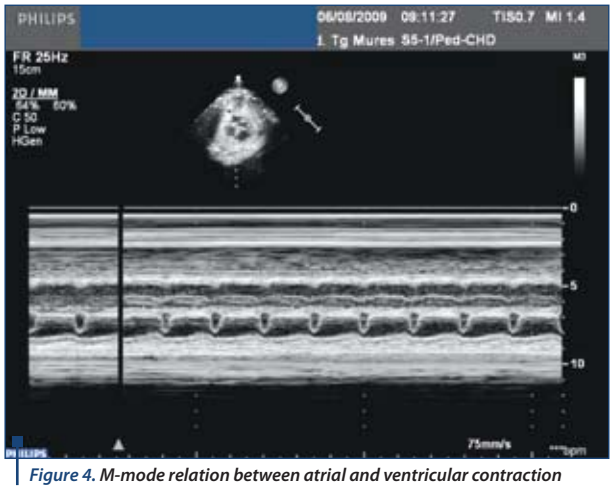

Figure 4. M-mode relation between atrial and ventricular contraction

Cardiac lesions resulting in an abnormal 4-camber view:

- ✓ Ebstein anomaly/dysplastic tricuspid valve;
- ✓ tricuspid/mitral atresia;
- ✓ atrioventricular complete and partial canal;
- ✓ hipoplastic left heart syndrom;
- ✓ pulmonary atresia with intact ventricular septum;
- ✓ double inlet left ventricle;
- ✓ corected transposition of the great arterias;
- ✓ large ventricular septal defect;
- ✓ cardiac tumors;
- ✓ severe aortic stenosis.

7. Extended views (including demonstration of outflow tracts):

- indentification of the aorta and pulmonary origins;
- charateristics of normal great arteries: one arising from each ventricle; the arteries cross each other soon after leaving the heart; the pulmonary arteria divides into 3 (each branch and the ductus);

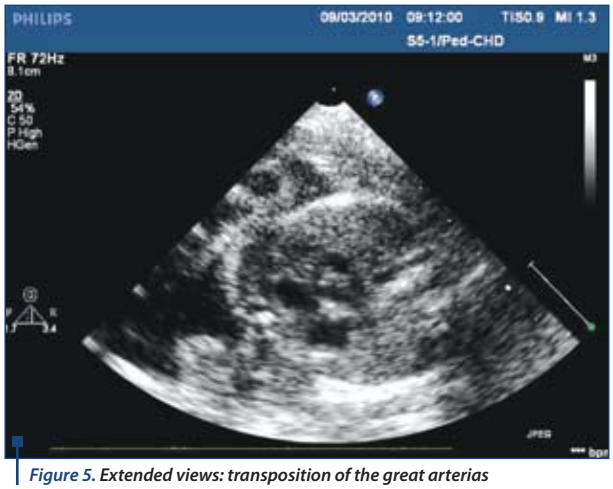

Figure 5. Extended views: transposition of the great arterias

- pulmonary artery continues as the ductus arteriosus and joins the proximal descendentic aorta;
- the aorta gives rise to head and neck braches;

- normally the pulmonary artery is slightly bigger than the aorta.

Cardiac lesions which can be detected using extended views:

- ✓ tetralogy of Fallot;
- ✓ pulmonary atresia with ventricular septal defect;
- ✓ double outlet right ventricle;
- ✓ transposition of the great arteries;
- ✓ truncus arteriosus;
- ✓ some coarctation;
- ✓ pulmonary stenosis.

8. The 3-vessel view

- pulmonary arteria is anterior to and bigger than;
- the aorta, which in turn is anterior to and bigger than;
- the superior vena cava.

5 transverse views:

1. transverse view of fetal abdomen to demonstrate situs;
2. 4-chamber view of heart;
3. 5-chamber view demonstrating aortic root arising from left ventricle;
4. bifurcation of the pulmonary artery;
5. 3 vessel view with tracheea at level of main PA joining the ductus arteriosus.

Identification of cardiac rhythm: M-mode echocardiography, placing cursor through an atrial wall and a ventricular wall simultaneously[9,13,20]. Supraventricular tachycardia and atrial flutter are the most frequently encountered fetal tachyarrhythmias. The presence of atrioventricular heart block permits the differential diagnosis with the atrial flutter[10,22,23].

Results

In the previously mentioned period they were examined a number of 45 pregnant, with the gestational age between 20 and 37 weeks (of gestation). The gestational age at the moment of the diagnosis and the conclusion of scanning the fetal heart are shown in table 1.

The reasons for which patients were examined in our service are given in table 2.

Of the total patients examined, 10 patients presented normal pregnancy. In 35 patients structural cardiac anomalies or heart rhythm disorders were found. The classification according to the fetal cardiac pathology is given in table 3.

Of the total 35 patients with cardiac pathology, 25 cases presented modifications of the 4-chamber section.

Of the total 45 patients examined, 21 cases were monitored in the postnatal period as well. Each neonate was examined by echocardiography immediately after birth by the pediatric cardiologist. The echocardiographic exam begins with the subxiphoidian view, very useful for the establishment of the abdominal and atrial situs, and of the veno-atrial, atrio-ventricular and ventriculo-arterial connections. From the parasternal long axis view the mitral valve, aortic valve and the left ventricle are visualized; in the apical 4 chamber view the atrioventricular valves are visualized. The relationship between the great arteries, the bifurcation of the pulmonary artery and the ductus arteriosus respectively can be visualized from the high parasternal view. The suprasternal view is useful for the visualization of the aortic arch[21]. After the establishment of the diagnosis and the evaluation of the hemodynamic status of the patient the surgical indication was established. The postnatal evolution of the monitored cases is given in table 4.

Discussion

Studies in specialized literature report a percentage of antenatal diagnosis of cardiac anomalies between 17.9 and 55.6 %[18]. Of 45 cases which were examined in our service in 35 cases, respectively 77.77%, was given the diagnosis of a fetal heart disease, congenital heart malformation or rhythm disorder. Most patients, respectively 39 out of 45, representing 86.66%, were examined in the pediatric cardiology service by the obstetrician's order, which detected a structural cardiac anomaly or a heart rhythm disorder. We consider that a good collaboration between the obstetrician and the pediatric cardiologist improves the understanding of hemodynamic changes in fetal heart malformation. Also, knowing the available treatment possibilities and the postnatal evolution, we are able to appreciate the prognosis of the neonate with congenital heart disease. And, because most newborns come from mothers with low risk[15], the determination of the antenatal diagnosis depends on a thorough and correct fetal screening.

The moment in which a complete scanning of the fetal heart can be performed is between 18 and 20 weeks of gestation. At 18 weeks all the atrioventricular connections, can be visualized, but sometimes minor injuries can not be excluded. From of the quality of the image, the point of view, the optimal period of examination is between 24 to 28 week[17,23]. In the studied group the gestational age at which the echocardiography of the fetus was performed in our service, was big; cases with the gestational age of over 29 weeks prevail.

The 4-chamber image is the most frequently used along the screening routine. It is in the same time the most important incidence, because over 60% of congenital heart malformations contain alterations of this image, on the condition that the view analyzed and obtained correctly[1,3]. Within the group of examined patients, out of 35 fetuses with heart malformation 25, respectively 71.42% had a modified 4-chamber image.

The diagnosed pathology is dominated by structural anomalies, 31 fetuses being with congenital heart malformations. In 4 cases emerged rhythm disorders: 2 cases of supraventricular tachycardia, 1 case of total atrioventricular block and 1 case of atrial extrasystoles. The cases of supraventricular tachycardia were correctly diagnosed in utero, based on the M mode aspect, where the relationship between the atrial and ventricular contraction was analyzed, the ventricu-

Table 1 Diagnosis of the cases

Crt. no.	Gestational age (weeks)	Diagnostic
1	30	Double outlet right ventricle. VSD. TGA. PS
2	32	Double outlet right ventricle. VSD. TGA. PS
3	34	Normal pregnancy
4	24	Pulmonary atresia with intact ventricular septum
5	22	Prematuar atrial beats
6	34	Coarctation of the aorta
7	33	Pulmonary atresia. VSD
8	32	Atrioventricular septal defect
9	29	Critical aortic stenosis
10	24	Tetralogy of Fallot
11	22	Normal pregnancy
12	36	SVT. Cardiomegaly. Mi I. TI
13	24	Normal pregnancy
14	26	Normal pregnancy
15	33	SVT. Cardiomegaly
16	33	Normal pregnancy
17	22	Severe aortic stenosis
18	36	Normal pregnancy
19	37	Normal pregnancy
20	22	Normal pregnancy
21	37	Heterotaxy syndrome
22	30	Double outlet single ventrile
23	21	TAC. Truncal valve stenosis
24	33	Hypoplastic left heart syndrome
25	31	Severe coarctation of the aorta. Aortic stenosis
26	22	Tricuspid valve dysplasia. Severe TI
27	24	Transposition of the great arteries
28	22	Tetralogy of Fallot. Hypoplastic PA
29	30	Total AVB. Hydrops fetalis
30	33	Double outlet right ventricle. VSD. TGA. PS
31	33	Pulmonary atresia. TI
32	27	Large inlet VSD . TI
33	23	Hypoplastic left heart syndrome
34	26	Atrioventricular septal defect
35	30	Single ventricle with undefined morphology. Malposition of the great arteries.
36	33	Hypoplastic left heart syndrome. VSD
37	33	Tricuspid atresia. PS
38	35	Normal pregnancy
39	37	Atrioventricular septal defect
40	22	Normal pregnancy
41	36	Coarctation of the aorta
42	34	Tricuspid valve dysplasia. Severe TI
43	33	Atrioventricular septal defect
44	36	Hypoplastic left heart physiopathology.
45	34	Mitral stenosis. Double outlet right ventricle. ASD. VSD

VSD ventricular septal defect, TGA transpositionof the great arteries, TAC trunckus arteriosus, TI tricuspid insufficiency, PS pulmonary stenosis, SVT supraventricular tachycardia, Mil mitral isufficiency, PA pulmonary artery, AVB atrioventricular block, ASD atrial septal defect

Table 2 The classification according to the motivation of the examination

Motivation of the examination	Number of pregnant women
Previous child or fetus with cardiac anomalies	5
Arrhythmias	5
Modification on screening exam	35

Table 3 The classification according to the fetal cardiac pathology

Fetal cardiac pathology	Type of affection and number of cases
Arrhythmias	4 SVT - 2 Total AVB - 1 Atrial extrasystolie - 1
The dominant congenital cardiac lesion	31 Single ventricle - 4 Hypoplastic left heart syndrome - 3 Subvalvular Ao stenosis. Coarctation of the aorta. VSD. - 1 Hypoplastic left heart syndrome with VSD - 1 Atrioventricular septal defect - 4 Double outlet right ventricle - 3 Pulmonary atresia - 3 Coarctation of the aorta - 3 Aortic stenosis - 2 Tetralogy of Fallot - 2 Tricuspid insufficiency - 2 Truncus arteriosus - 1 Transposition of the great arteries - 1 Ventricular septal defect - 1 Heterotaxy syndrome - 1

SVT supraventricular tachycardia, AVB atrioventricular block, VSD ventricular septal defect, Ao aorta

lo-atrial interval being short[13,26]. Newborns needed cardiac anti-arrhythmic and anti-congestive treatment immediately after birth, because the supraventricular arrhythmia, although it has been diagnosed during fetal life, it was not controlled. Congenital heart malformation were diagnosed in 31 cases (68.88%) among them the complex heart malformations (single ventricle, hypoplastic left heart syndrome, double outlet right ventricle) were the most frequent. Conotruncal defects, characterized by anomalies in the conotruncal septum represent an important group of heart disease[14]. In the study group we had a number of 7 cases of conotruncale anomalies. 21 fetuses were monitored in the postnatal period, too. The antenatal diagnosis was confirmed in all cases and 15 cases benefited from surgical intervention. In the case of the newborn in whom prenatal diagnosis of hypoplastic left heart physiopathology was made, without the possibility of being classified in the hypoplastic left heart syndrome group of disease, after 3 days of life the diagnosis of subvalvular aortic stenosis, ventricular septal defect and coarctation of the aorta was established. The antenatal diagnosis allows the determination of a therapeutic strategy in the case of severe damage, which could go undiagnosed in the immediate neonatal period[12,24]. One of the most important congenital heart disease in which the a improved survival and reduced morbidity is coarctation of the aorta, a ductal dependent lesion, which evolves postnatally with hemodynamic instability and neurologic complications[7]. In the 3 cases of coarctation of the aorta diagnosed in our group of study, treatment was instituted immediately after birth with prostaglandin to maintain patency of ductus arteriosus, and subsequently were operated, having a very good postoperative evolution.

Conclusions

The improvement of the antenatal diagnosis of heart diseases can be achieved with fetal echocardiographic screening based on the main incidences of visualization of the fetal heart, at an optimal age of pregnancy.

Cooperation between the obstetrician and the pediatric cardiologist increases accuracy of antenatal diagnosis.

A correct antenatal diagnosis allows the information of parents as accurate as possible, regarding the heart diagnosis of the fetus and the elaboration of the most efficient therapeutic strategy. ■

Table 4 The postnatal evolution of the monitored cases

Type of congenital heart disease	Postnatal evolution
Double outlet right ventricle. VSD. TGA.	Corrective surgical treatment
Coarctation of the aorta	Corrective surgical treatment
Atrioventricular septal defect	Corrective surgical treatment
SVT. Cardiomegaly. Mi I. TI	Birth at 36 weeks, antiarrhythmic treatment
SVT. Cardiomegaly	Birth at 37 weeks, antiarrhythmic treatment
Heterotaxy syndrome	Postnatal echocardiography
Truncus arteriosus	Corrective surgical treatment
Pulmonary atresia	Surgical treatment
Atrioventricular septal defect	Periodical evaluations
Transposition of the great arteries	Corrective surgical treatment
Single ventricle. Transposition of the great arteries	Surgical treatment
Hypoplastic left heart syndrome	Neonatal death
Tricuspid atresia. Pulmonary stenosis	Surgical treatment
Coactation of the aorta	Surgical treatment
Pulmonary atresia. VSD	Surgical treatment
Hypoplastic left heart syndrome	Neonatal death
Subvalvular aortic stenosis. CoAo. VSD	Surgical treatment
Tricuspid valve dysplasia. Severe TI	Surgical treatment
Single ventricle. Pulmonary stenosis.	Surgical treatment
Atrioventricular septal defect	Surgical treatment
Single ventricle. TGA. Pulmonary stenosis. Mitral stenosis.	Surgical treatment

References

1. Abu-Rustum R.S., Daou L., Abu-Rustum S.E., Role of ultrasonography in early gestation in the diagnosis of congenital heart defects, J Ultrasound Med, 2010; 29: 817-21.
2. Allan L., Technique of fetal echocardiography, Pediatr Cardiol, 2004; 25: 223-33.
3. Archer N., Manning N., Fetal Cardiology, Oxford University Press, 2009; 47-66.
4. Dolk H., Loane M., Garne E., The prevalence of congenital anomalies in Europe, Adv Exp Med Biol, 2010; 686: 349-64.
5. Eronen M., Outcome of fetuses with heart disease diagnosed in utero, Arch Dis Child, 1997; 77: 41-6.
6. Ferencz C., Rubin J.D., McCarter R.J. et al., Congenital heart disease: prevalence at live. The Baltimore-Washington Infant Study, Am J Epidemiol, 1985; 121: 31/36.
7. Franklin O., Burch M., Manning N., Prenatal diagnosis of corctation of the aorta improves survival and reduces morbidity, Heart, 2002; 87: 67-9.
8. Hui L., Wei J., Ying M. et al., Prenatal diagnosis of congenital fetal heart abnormalities and clinical analysis, Zhejiang Univ SCI, 2005; 6B (9): 903-6.
9. Ito S., Magee L., Smallhorn J., Drug therapy for fetal arrhythmias, Clin Perinatol 1994; 21: 543-73.
10. Krapp M., Kohl T., Simpson J.M. et al., Review of diagnosis, treatment and outcome of fetal atrial flutter compared with supraventricular tachycardia, Heart, 2003; 89:913-7.
11. Lee W., L. Allan, J.S. Carvalho et al. ISUOG consensus statement: what constitutes a fetal echocardiogram? Ultrasound Obstet Gynecol 2008; 32: 239-242.
12. Lagopoulos M.E., Manlhiot C., McCrindle B.W., Jaeggi E.T. et al., Impact of prenatal diagnosis and anatomical subtype on outcome in double outlet right ventricle, Am Heart J, 2010; 160: 692-700.
13. Maragnes P., Fournier A., Lessard M. et al., Evaluation and prognostic of fetal arrhythmia, Pediatrie, 1991; 46: 481-8.
14. Markov D., Fetal echocardiography: part III- the conotruncal anomalies, Akush Ginekol, 2010; 49: 25-33.
15. Ozkutlu S., Akca T., kafai G. et al., The results of fetal echocardiography in a tertiary center and comparison of low- and high- risk pregnancies for fetal congenital heart defects, Anadolu kardiyol Derg, 2010; 10:263-9.
16. Shima Y., Shindoh F., Nakajima M., Prenatal diagnosis of congenital heart disease: clinical experience and analysis, J Nippon Med Sch, 2004; 71: 328-31.
17. Small M., Copel J.A., Indications for fetal echocardiography, Pediatr Cardiol, 2004; 25: 210-22.
18. Stoll C., Garne E., Clementi M., Evaluation of prenatal diagnosis of associated congenital heart diseases by fetal ultrasonographic examination in Europe, Prenat Diadn Vol 21 Issue 4Apr 2001: 242-53.
19. Sturla E.N., Lee W., Carvalho J.S., Chaoui R., Copel J., Hecher K., Paladini D., Cardiac screening examination of the fetus: guidelines for performing the basic and extended basic scan. Ultrasound Obstet Gynecol, 2006; 27: 107-13.
20. Van Engelean A., Weijtens O., Brenner J.I. et al., Management outcome and follow-up of fetal tachycardia, J Am Coll Cardiol 1994; 24: 1371-5.
21. Wood D., Respondek-Liberska M., Puerto B. et al., Perinatal echocardiography: protocols for evaluating the fetal and neonatal heart. J. Perinat. Med. 2009, 37:5-11.
22. Wren C., Mechanisms of fetal tachycardia, Heart, 1998; 79: 536-7.
23. Wu T.H., Huang L.C., Ho M., fetal atrial flutter: a case report and experience of sotalol treatment, J Obstet Gynecol 2006.
24. Yagel S., Silverman N.H., Gembruch U., Fetal cardiology, Taylor&Francis, 2003; 335-55.
25. Yang S., Zhu Q., Chen J. et al., Echocardiography diagnosis of fetal cardiac structural abnormalities, Zhongguo Dang Dai Er Ke Za Zhi, 2010; 12: 99-102.
26. Zhao B.W., Pan M., Zhang S.Y. et al., Application of fetal echocardiography in detection of fetal arrhythmia and its clinical significance, Zhonghua Fu Chan Ke Za Zhi, 2004; 39: 365-8.