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Fetal echocardiography in Lithuania: traditions, significance and problems

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Background. The discovery of ultrasound has made a revolution in almost all fields of medicine. The past three decades have withessed an intensive development of fetal echocardiography methods and technique. The aim of the paper is to present a review of the results and trends of the last 10 years of fetal echocardiography in Lithuania and to show the spectrum and outcomes of prenatally detected congenital heart diseases.

Materials and methods. Fetal echocardiography was performed for 1816 fetuses during the period from 1999 to 2009.

Results. Cardiac pathology was diagnosed in 176 (9.7%) fetuses. Heart defects were detected in 112 (63.6%) of them, cardiac rhythm and conduction disturbances in 62 (35.2%), cardiomyopathy in 2 (1.1%) fetuses, and heart rhabdomyoma in 1 (0.6%) fetus. The general rate of the postnatal diagnosis of congenital heart defects in Lithuania was about 10%. Most of fetal cardiac diseases (70.5%) were diagnosed after 22 weeks of gestation. Because most of antenatally diagnosed congenital heart defects (74%) were critical and inconsistent with life, a large part of newborns (40.2%) died in the neonatal period, 10.7% of fetuses died *in utero*, and 8% of pregnancies were terminated by abortion. The data demonstrate good tendencies: the diagnosis has become earlier, a wider spectrum of diseases have been diagnosed, more newborns have survived. Our survey shows that 41.1% of newborns with prenatally diagnosed congenital heart defects have survived.

Conclusions. 10% of severe congenital heart diseases are detected prenatally in Lithuania. The efficacy of antenatal diagnostics depends on the qualification of specialists, the number of tertiary care centers, on a successful collaboration among pediatric cardiologists, obstetricians and geneticists. The main problem is an insufficient preparation of obstetricians, the uncertified favor of pediatric cardiologist.

Key words: congenital heart disease, fetal echocardiography, antenatal diagnostics

INTRODUCTION

Modern medical technology development, effective diagnostic devices and the emergence of new diagnostic methods have made a revolution in medicine. Ultrasound diagnosis has passed several stages of the development over 20–30 years and reached amazing achievements hardly imaginable three decades ago. The possibility of diagnosing of congenital cardiac pathology before child's birth was first considered in the early 1980s when fetal echocardiography was developed and implemented in fetal ultrasound techniques (1, 2). Before the 1990, such complex heart defects as coarctation of the aorta, tetralogy of Fallot, *truncus arteriosus*, total anomalous pulmonary venous connection and other defects of the fetus heart were described in the scientific literature. Recently, modern complex, informative, non-invasive, harmless fetal cardiac ultrasound assay methods have become very important in evaluating and diagnosing various cardiac pathological changes. The most important fetal and neonatal echocardiography objectives are to diagnose congenital heart defects and rhythm disturbances, to assess hemodynamic changes under various physiological and pathological conditions of the fetus (3).

Congenital heart defects are the most common fetal anomalies. Lithuania is a country of 3 339.4 thousand people with 36 651 live births in 2009 (4). In many countries, including Lithuania, congenital heart defects occur in 6–10 newborns per 1 000 live births. This pathology is about 6.5 times more frequent than chromosomal abnormalities and about 4

times more frequent than neural tube defects (5). About one third of heart defects (3–4 newborns per 1 000 live births) are critical and inconsistent with live as a newborn needs immediate surgery and drug therapy. In Lithuania, critical congenital heart defects are diagnosed for about 100 newborns every year. About 20–30 infants are operated on in the first month of life.

Fetal echocardiography is still a difficult examination. The investigators' experience, the quality of ultrasound equipment, maternal obesity and maternal abdominal wall thickness, oligohydramnion and sometimes fetal position and active fetal movements are the factors affecting the interpretation of images (6). The nature of the cardiac anatomy and the wide spectrum of defects make the heart examination quite difficult.

Indications for fetal echocardiography (7, 8) are the following:

1) maternal risk factors: maternal or familial heart disease (congenital heart defect, cardiomyopathy), exposure to teratogens (alcohol, valproic acid, lithium), metabolic disorders (type 1 or 2 diabetes mellitus, phenylketonuria), maternal autoimmune disease, infectious diseases (Rubella infection), *in vitro* fertilization (the prevalence of congenital heart defects increases up to 3-fold);

2) fetal risk factors: obstetric ultrasound suggesting fetal heart disease, risk for fetal cardiovascular compromise (twin–twin transfusion syndrome, fetal arteriovenous malformations, fetal anemia), obstetric ultrasound suggesting fetal extracardiac disease (other organ anomalies – combined development abnormalities of several organs occur in 25–45% of cases, chromosomal abnormalities, increased nuchal translucency (more than 3,5 mm presenting at 10–13 weeks of gestation even in the absence of chromosomal anomaly), fetal arrhythmias, non-immune fetal hydrops, fetal intrauterine growth retardation.

However, about 80% of infants with congenital heart pathology usually are born to mothers without known risk factors, so the screening at 18–22 weeks of gestation and measuring nuchal translucency in the early period of pregnancy are very important (9).

Many of fetal heart defects can be identified by assessing the standard images of a normal fetal heart at a routine fetal scan (10). A more detailed assessment of cardiac defects and formulation of a precise statement to parents must be made by a professional – a pediatric cardiologist with expertise in fetal and pediatric echocardiography. A complete fetal heart examination includes two-dimensional echocardiography, color and spectral Doppler, M-mode echocardiography. 3D, 4D mode echocardiography has also been applied recently (11).

MATERIALS AND METHODS

During 1999–2009, fetal echocardiography was performed in 1816 fetuses at Vilnius University Children's Hospital. The most common indications for fetal echocardiography were the suspect of fetal cardiac anomaly found at a routine fetal scan, fetal heart rhythm disturbances, and a family history of congenital heart disease. The heart condition of fetuses was assessed at the age of 16–40 weeks of gestation.

Cardiac structure and vascular anatomy were evaluated using the standard ultrasound imaging. In the standard fetal cardiac 4-chamber view (Fig. 1), were determined fetal cardiac axis and position (normal angle rate is about 40°), size (normal heart occupies approximately 1/3 of the chest, the cardio-thoracic ratio represents about 50%), heart chambers' proportion (normally left and right chambers are of approximately equal size, with the right ventricle becoming slightly larger than the left ventricle as the pregnancy progresses). Heart function (measured as a shortening fraction >30%) and rhythm were also measured.

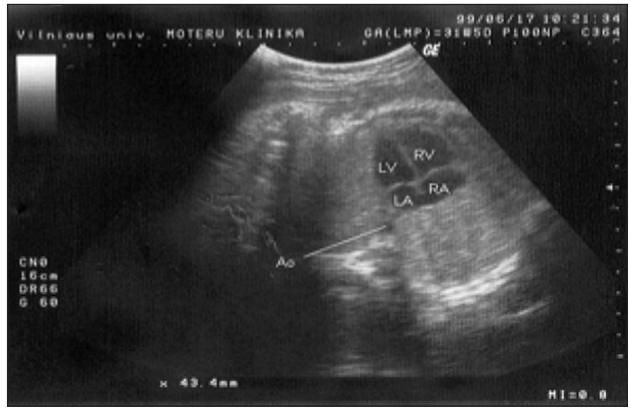

Fig. 1. The four-chamber view (LV – left ventricle, RV – right ventricle, LA – left atrium, RA – right ventricle, Ao – aorta)

The other images used to assess vascular pathology were the left ventricular outflow tract – aorta (Fig. 2), the right ventricular outflow tract – pulmonary artery (Fig. 3), the three-vessel view (Fig. 4), the aortic arch (Fig. 5) and *ductus arteriosus* (Fig. 6).

Fetal cardiac pathology was diagnosed on the basis of the following ultrasound characteristics:

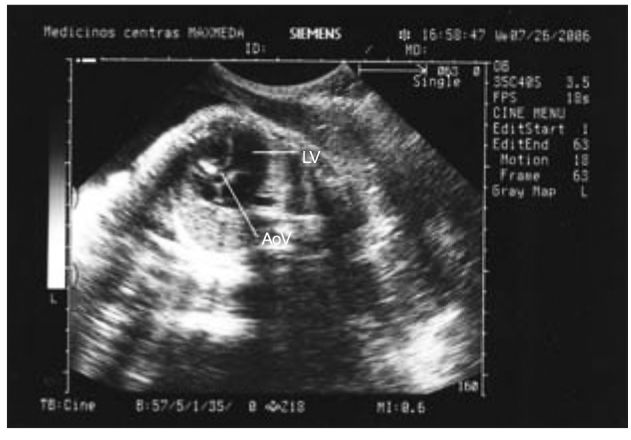

Fig. 2. The left ventricular outflow tract (LV – left ventricle, AoV – aortic valve)

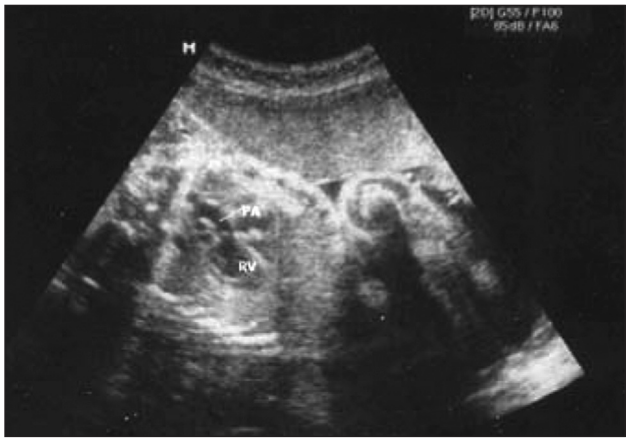

Fig. 3. The right ventricle (RV – right ventricle, PA – pulmonary artery)

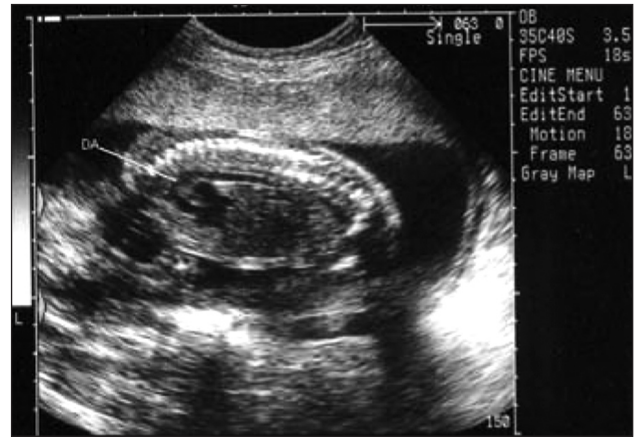

Fig. 6. Ductus arteriosus (DA)

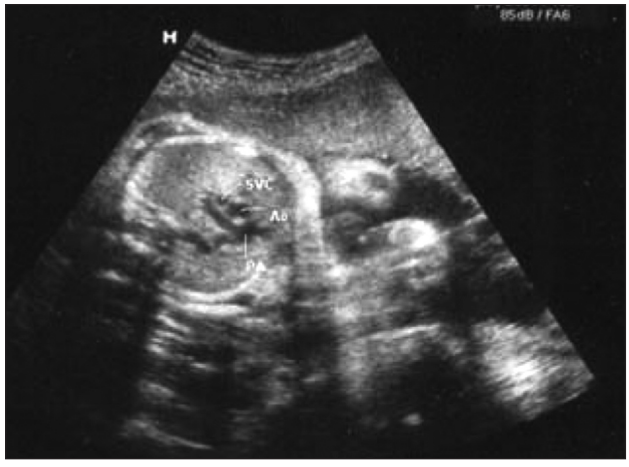

Fig. 4. The three-vessel view (SVC – superior vena cava, Ao – aorta, PA – pulmonary artery)

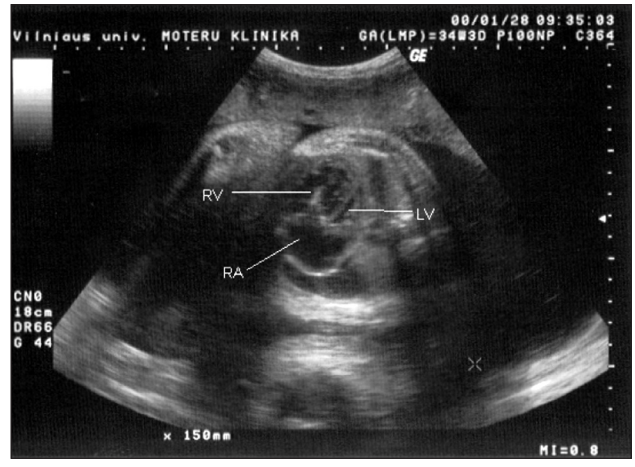

Fig. 7. Altered cardiac axis (RV – right ventricle, LV – left ventricle, RA – right atrium)

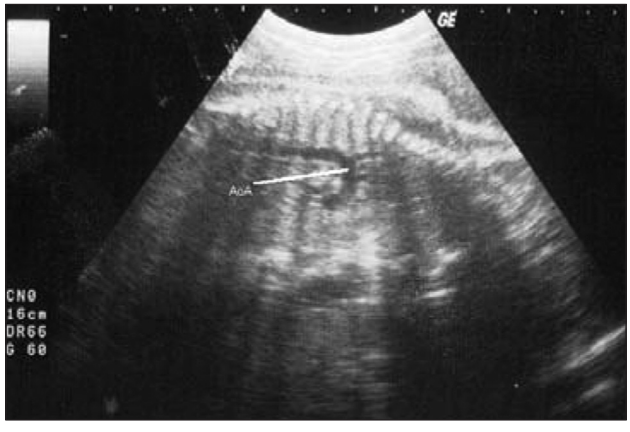

Fig. 5. Aortic arch (AoA)

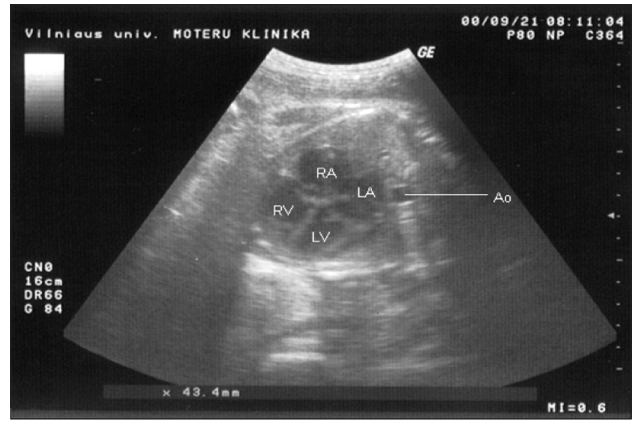

Fig. 8. Cardiomegaly (LV – left ventricle, RV – right ventricle, RA – right atrium, LA – left atrium, Ao – aorta)

1) cardiac malposition (dextrocardia, with or without *situs inversus*, mesocardia, *situs ambiquous*).

2) altered cardiac axis (Fig. 7). In 40–45% of fetal cardiac anomalies, the fetal cardiac axis is deviated more than 57°; when the cardiac axis deviation exceeds 75°, the probability of heart defect is about 70–80%;

3) cardiomegaly (Fig. 8). In most cases, cardiomegaly is caused by fetal cardiac pathology (heart defect, cardiomyopathy, abnormal heart rhythm, atrioventricular valve regurgitation). Other reasons are a relatively small chest or chest compression (in case of bone dysplasia, oligohydramnion), dynamic fetal blood flow (fetal anemia), fetal hypoxia, maternal diabetes;

4) disproportion of the cardiac chambers, dilatation of the right-heart or left-heart chambers. The most common

cause of the right heart enlargement (RV/LV > 1.24) are coarctation of the aorta and other heart defects (pulmonary trunk atresia, anomalous pulmonary venous drainage, left heart hypoplasia), also tricuspid valve regurgitation, intracranial *v. Galenae* aneurysm, intrauterine growth retardation (impaired fetal and placental blood flow), and dynamic fetal blood flow. The left heart enlargement (RV/LV < 0.8) is most common in case of other heart defects (aortic stenosis, tricuspid valve atresia) and cardiomyopathy.

5) changes of the cardiac and vascular structure. Ventricular septal defect, a complete atrioventricular septal defect, valve abnormalities (tricuspid atresia, Ebstein's anomaly), common arterial trunk, aortic stenosis or atresia, tetralogy of Fallot, hypoplastic left heart syndrome occur most frequently;

6) changes of the cardiac ventricular function under investigation (a decrease of myocardial contraction and shortened fraction, dilatation of the right atrium, tricuspid valve regurgitation);

7) pericardial effusion (more than 3–4 mm);

8) arrhythmias.

RESULTS

Among 1 816 fetuses, cardiac pathology was diagnosed in 176 (9.7%). Of them, 112 (63.6%) were heart defects, 62 (35.2%) cardiac rhythm and conduction disturbances, 2 (1.1%) cardiomyopathies, 1 (0.6%) heart rhabdomyoma. In 65 fetuses, other organ anomalies were detected (without cardiac defect); 1 575 fetuses were developing normally.

A wide spectrum of congenital heart defects (septal defects – Fig. 9, aortic stenosis – Fig. 10, tetralogy of Fallot, pulmonary artery atresia or stenosis – Fig. 11, tricuspid valve atresia – Fig. 12, hypoplastic left heart syndrome – Fig. 13 and other complex heart diseases) were diagnosed antenatally. Most of fetal cardiac defects (70.5%) were diagnosed after 22 weeks of gestation; 17% of fetal cardiac defects (19 fetuses) were associated with chromosomal anomalies, and 17.8% of the defects (23 fetuses) were associated with other organ anomalies. Chromosomal and genetic disorders were associated with a ventricular septal defect, atrioventricular septal defect, tetralogy of Fallot, pulmonary artery stenosis. Isolated congenital heart defects were diagnosed in 93 fetuses (83%).

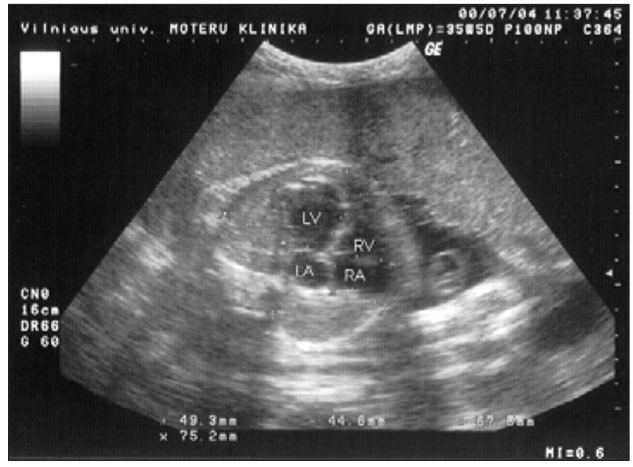

Fig. 10. Aortic stenosis

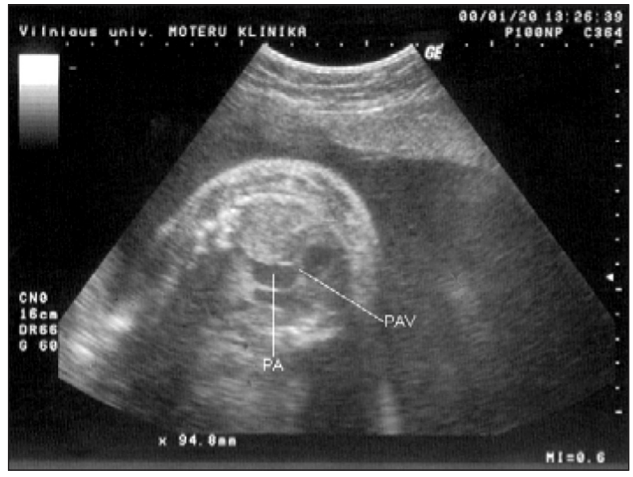

Fig. 11. Pulmonary valve stenosis (PA – pulmonary artery, PAV – pulmonary valve)

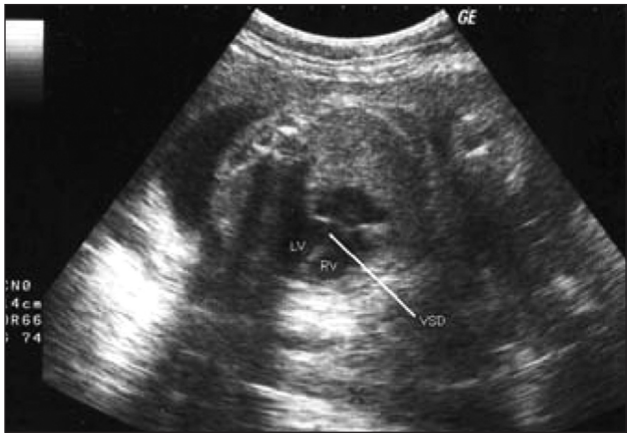

Fig. 9. Ventricular septal defect (LV – left ventricle, RV – right ventricle, VSD – ventricular septal defect)

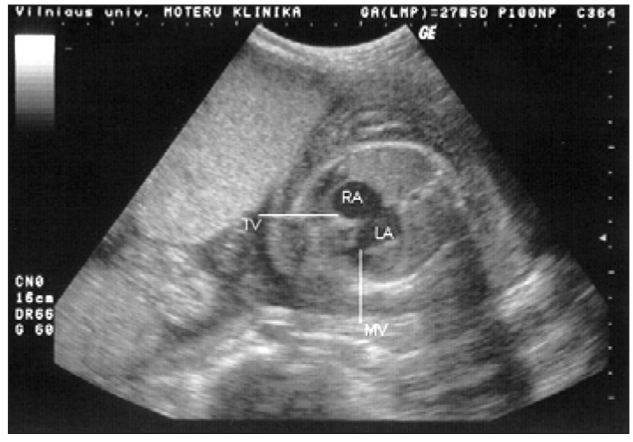

Fig. 12. Tricuspid valve atresia (TV – tricuspid valve, MV – mitral valve, RA – right atrium, LA – left atrium)

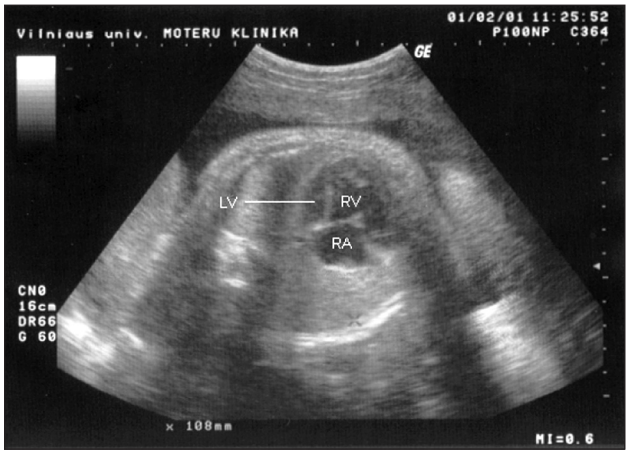

Fig. 13. Hypoplastic left heart syndrome

After the discussion and consensus of obstetrician, geneticist and pediatric cardiologist, the termination of pregnancy was proposed to parents in cases of hypoplastic left heart syndrome (4 cases), in aortic stenosis (2 cases), in combined cardiac defects with extracardiac anomalies (3 cases).

In the most cases of fetal cardiac pathology, no significant hemodynamic disturbances were determined because of fetal circulation peculiarities. The general condition of most fetuses was good as it was compensated by the functional *foramen ovale, ductus arteriosus* and other mechanisms. Two fetuses had a complete heart block with a cardiac defect, and in one fetus supraventricular tachycardia developed.

Our observation period (1999–2009) was separated into two periods – 1999–2004 and 2005–2009, because fetal cardiac examination started in 2000–2002 and the antenatal diagnostics of congenital heart diseases improved in 2005–2009. We compared the tendencies of fetal echocardiography and the survival rate of the examined fetuses. The results are shown in Table 1.

The quantitative analysis of echocardiographic examination in Lithuania over 1999–2004 and 2005–2009 shows that the number of consulted women increased, but the number of antenatally diagnosed cardiac defects decreased. The majority of fetuses examined in 2005–2009 were healthy, thus, many of the requests for echocardiographic examination were unreasonable, reflecting a poor situation in the primary care. Cardiac pathology was suspected frequently without evidence during a routine obstetric ultrasound examination, i. e. obstetricians have too little knowledge and experience of fetal heart anatomy and ultrasound views. Our data demonstrate also good tendencies: more cardiac defects were diagnosed before 22 weeks of gestation (19.6% in 1999–2004 versus 36.4% in 2005–2009), the number of late antenatal diagnosis cases were diminishing (80.4% versus 63.6%), more newborns survived (30.4% versus 48.5%), the mortality of newborns and fetuses decreased. The survival diagram of newborns with antenatally diagnosed heart defects over 1999–2009 is shown in Fig. 14. The survival decreased in 2001–2004 and was gradually increasing in 2005–2009 because of improved surgical care and intensive therapy facilities.

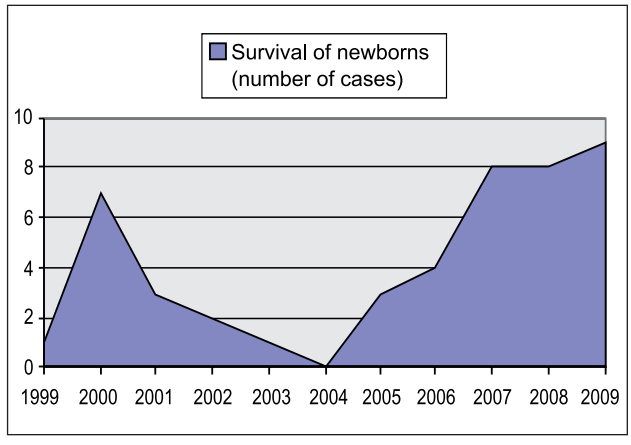

Fig. 14. Survival of newborns with antenatally diagnosed heart defects in 1999–2009

Table 1. **Quantitative analysis of echocardiographic examination in Lithuania over 1999–2004 and 2005–2009**

	1999–2004	2005–2009	1999–2009
Number of examined fetuses	446	1370	1816
Number of cardiac defects	46 (10.3%)	66 (4.8%)	112 (6.2%)
	< 0.0001 (Fisher); p < 0.0001, Chi square = 17.565		
Diagnostics before 22 weeks	9 (19.6%)	24 (36.4%)	33 (29.5%)
	0.0614 (F). p = 0.0550. Chi square = 3.681		
Diagnostics after 22 weeks	37 (80.4%)	42 (63.6%)	79 (70.5%)
	0.0614 (F). p = 0.0550. Chi square = 3.681		
Rate of survival	14 (30.4%)	32 (48.5%)	46 (41.1%)
	0.0786 (F). p = 0.0561. Chi square = 3.649		
Rate of deaths after birth	23 (50%)	22 (33%)	45 (40.2%)
	0.0826 (F). p = 0.0767. Chi square = 3.133		
Termination of pregnancy	2 (4.3%)	7 (10.6%)	9 (8%)
	0.3038 (F). p = 0.2307. Chi square = 1.437		
Rate of intrauterine deaths	7 (15.2%)	5 (7.6%)	12 (10.7%)
	0.2262 (F). p = 0.1983. Chi square = 1.655		

Because most of antenatally diagnosed congenital heart defects (55.3%) were critical and inconsistent with life, a big part of newborns (40.2%) died in the neonatal period, 10.7% of fetuses died *in utero*, and 8% of pregnancies were terminated by abortion.

DISCUSSION

Fetal cardiac ultrasound testing in Lithuania was started in 1998–1999. Antenatal diagnostics of the complex heart defects with high visibile signs in a fetal cardiac 4-chamber view (large septal defects, left heart hypoplasia, atresia of the pulmonary artery and the tricuspid valve, etc.) were being performed. The training of obstetricians, gynecologists and ultrasound specialists in the fetal cardiac ultrasound examination methodology started in 2000–2002, but a very small number of fetal heart defects were diagnosed before 2005. The majority of congenital heart diseases were diagnosed antenatally after 22 weeks of gestation (the mean gestational age at the time of the diagnosis was 31 weeks). The prognosis of these heart defects was usually poor.

The antenatal diagnostics of congenital heart diseases improved in 2005–2009 because of persistent theoretical and practical work in this area, the increased rate of suspicion of pathology during routine fetal ultrasound analysis, and the growing interest in this area. A larger number of cardiac defects were detected during this period. Over the recent years, more cases of aortic stenosis, complete atrioventricular septal defect, Ebstein's anomaly, pulmonary atresia and stenosis, the first cases of transposition of the great arteries have been diagnosed. More diagnoses were made before 22 weeks of gestation in comparison with the period 1999–2004. Over the past five years, the survival rate has increased (from 30.4% to 48.5%) and the number of neonatal deaths has decreased (50% versus 33%).

In Lithuania, if fetal cardiac pathology is suspected during obstetric scan, a woman is referred to the pediatric cardiologist. This favor is not yet certified by the State Patient's Fund as a routine observational schedule of pregnant women and is still unpaid. Also, only one pediatric cardiologist has enough experience in fetal echocardiography in the whole country. If a fetal cardiac pathology is confirmed, the woman is being sent for delivery to the Vilnius Perinatology Center. Most of fetal cardiac pathologies are concentrated in this Center.

During the period 1999–2009, echocardiography was performed postnatally in 8 845 newborns at Vilnius University Children's Hospital. Various congenital heart diseases were detected in 992 (10.4%) newborns; 216 (21.8%) of these heart defects were critical. These findings are similar to data on the antenatal diagnostics of congenital heart diseases: 9.7% of the examined fetuses had congenital heart defects ($p < 0.01$). These data show that, diagnosed either antenatally or postnatally, heart diseases are present in about 10% of babies (9.7% were diagnosed *in utero* and 10.4% after birth), so almost all heart defects can be detected before birth. Keeping in mind that some of extremely critical heart defects are inconsistent with life, early antenatal diagnosing would be a reason for the termination of pregnancy, thus saving the time and health of the mother and diminishing the costs of postnatal treatment.

The prenatal detection rate of congenital heart diseases remains low in most European countries, and a substantial proportion of infants with serious heart diseases are diagnosed only after discharge from hospital after birth. For example, the incidence of major congenital heart defects in Northern Norway is 4.4 per 1 000 births, the prenatal detection rate being 24%; 39% of cases were diagnosed postnatally [12]. In South Australia, congenital heart defects were present in 10.7 per 1 000 births, and of these only 22.5% were referred to fetal echocardiography (13). In Lithuania's neighbour-country Latvia, fetal cardiology started developing at the end of 1997 (14). A retrospective detailed clinical study including 225 echocardiograms of pathological findings was made in Latvia. Pathological findings were present in 9.2% of the total 2 445 examinations. Fetal heart pathology was diagnosed in 31% of cases up to the 22nd week of gestation. The prenatal detection rate of congenital heart defects in some countries such as France, UK is high (15, 16). The differences are shown in Table 2.

Table 2. **The incidence and prenatal diagnostics rate of congenital heart defects (CHD) in different countries**

Country	CHD incidence	CHD prenatal detection rate
France	5–6 per 1 000 births	53.7%
UK	5–7 per 1 000 births	52.8%
North Norway	4.4 per 1 000 births	24%
South Australia	10.7 per 1 000 births	22.5%
Latvia	ND	9.2%
Lithuania	6–8 per 1 000 births	6.2%

Infant survival depends on early diagnostics and treatment. It particularly suits for babies with the arterial duct dependent diseases. The proper antenatal diagnostics of heart defects would allow a sick baby to be born in a perinatology center, to access a cardiac surgical center in a stable condition and to be operated on as soon as possible. It also would help to prepare parents for a sick newborn birth and subsequent psychological difficulties.

The newborn survival rate in Lithuania has also changed over the last decade. Because of the significantly improved cardiac surgery assistance, more and more newborns with extreme cardiac defects survive: 75% of newborns and 92% of infants survived during the first year of life after cardiac surgery in 2009.

CONCLUSIONS

About 10% of severe congenital heart diseases are detected prenatally in Lithuania. The efficacy of antenatal diagnostics depends on the qualification of specialists, the number of tertiary care centers, on a successful collaboration among

pediatric cardiologists, obstetricians and geneticists. The main problems are the insufficient preparation of obstetricians and the uncertified favour of pediatric cardiologist.

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Ramunė Vankevičienė

VAISIAUS ECHOKARDIOGRAFIJA LIETUVOJE: TRADICIJOS, REIKŠMĖ IR PROBLEMOS

S a n t r a u k a

Įvadas. Ultragarso atsiradimas padarė perversmą visose medicinos srityse. Perėjusi kelis raidos etapus ultragarsinė diagnostika pasiekė sunkiai prieš 30 metų įsivaizduojamų laimėjimų. Šio straipsnio tikslas – apžvelgti vaisiaus echokardiografijos pasiekimus ir rezultatus Lietuvoje, aptarti antenataliai nustatytų įgimtų širdies ydų spektrą ir išeitis per 10 metų.

Medžiaga ir metodika. 1999–2009 m. buvo atliktos 1 816 vaisių echokardiografijos.

Rezultatai. Širdies patologija diagnozuota 176 (9,7 %) iš 1 816 tirtų vaisių. Iš jų širdies ydos buvo nustatytos 112 (63,6 %) vaisių, širdies ritmo ir laidumo sutrikimai – 62 (35,2 %), kardiomiopatijos – 2 (1,1 %) vaisiams, rabdomiomos – 1 vaisiui (0,6 %). Daugiausia vaisiaus širdies ydų (70,5 %) diagnozuota po 22 nėštumo savaitės. Kadangi dauguma (74 %) antenataliai diagnozuotų įgimtų širdies ydų buvo kritinės, nesuderinamos su gyvybe, didžioji dalis naujagimių (40,2 %) mirė naujagimystės periodu, 10,7 % vaisių žuvo iki gimimo, 8 % nėštumų buvo nutraukta. Gauti rezultatai rodo geras tendencijas: anksčiau nustatoma diagnozė, diagnozuojamas platesnis ligų spektras, daugiau išgyvena naujagimių. Antenataliai diagnozavus širdies patologiją išgyveno 41,1 % naujagimių.

Išvados. Lietuvoje iki gimimo nustatoma 10 % kritinių širdies ydų. Antenatalinės diagnostikos efektyvumas priklauso nuo specialistų kvalifikacijos, tretinio lygio centrų skaičiaus, glaudaus įvairių sričių specialistų (vaikų kardiologo, akušerio ginekologo ir genetiko) bendradarbiavimo. Tačiau vis dar nepakankamai parengiama pirminės grandies specialistų, neįteisinta vaikų kardiologo konsultacijos paslauga.

Raktažodžiai: įgimtos širdies ydos, vaisiaus echokardiografija, antenatalinė diagnostika