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EARLY PRENATAL ULTRASOUND DIAGNOSIS OF VENTRICULAR SEPTAL DEFECT

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Abstract
Ventricular septal defect (VSD) is a deficiency of the septum separating the ventricles. Usually, ventricular septal defects are correctly diagnosed when their dimensions exceed 4 mm, however, diagnosis of smaller defects is also possible. Various congenital heart defects were reported in 182 fetuses/newborns, among which breast cancer accounted for 25.8% (+3.2) or 47 cases. The age of the patients in the study group ranged from 18 to 40 years old. Early prenatal diagnosis was possible in 11 (23.4%+6.2) fetuses between 11 weeks of one day and 13 weeks of six days of pregnancy and was performed according to generally accepted diagnostic criteria. In the second half of pregnancy, DMF was additionally diagnosed in 7 (14.9%+5.2) fetuses. In the group with undiagnosed prenatal congenital heart defects, breast cancer was registered in 29 (61.7%+7.1) newborns. Various echographic markers and their combination were registered in 10 out of 11 (90.9%+8.7) fetuses with breast cancer diagnosed in early pregnancy. A thorough study of the ultrasound anatomy of the fetus made it possible to detect various extracardial abnormalities in 6 out of 11 (54.5%+15.0) fetuses at an early date. The outcome of pregnancy in the study group of fetuses was more often marked as favorable. Pregnancy ended with urgent delivery and the birth of live children in 37 (78.8%+6.0) patients.

Keywords: fetus, congenital heart disease, ventricular septal defect, transvaginal echocardiography, early prenatal diagnosis.

Introduction

Congenital heart defects (CHD) attract the attention of researchers all over the world not only because of their high incidence, but also because they cause 40% of perinatal losses and lead to deaths in the first year of life [1]. According to the Center for Disease Control (USA), it is CHD that most often, compared with other developmental abnormalities, lead to the death of children under one year of age [6]. The data of Russian specialists completely coincide with the data of their foreign colleagues. Despite the widespread introduction of prenatal ultrasound examinations, perinatal mortality from congenital malformations is steadily increasing and firmly holds the second place in the structure of perinatal losses. At the same time, multiple malformations account for 45.2%, and isolated CHD - 21.4%. For comparison, defects of the central nervous system

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account for only 14.3% (3rd place), and malformations of the gastrointestinal tract - 11.9% (4th place) [2]. High rates of death of children from CHD both in Russia and around the world are associated with difficulties in prenatal diagnosis of these defects. The entire history of the world prenatal echography proves that the diagnosis of any congenital malformation is based on a well-organized screening system for pregnant women. The availability of a state screening program, the clear interaction of specialists at the I, II and III levels of examination, the maintenance of national registers of congenital and hereditary pathology — all these factors significantly affect the quality of prenatal diagnosis and, in particular, the diagnosis of CHD in the fetus. On average, in different countries, the sensitivity of echography in the diagnosis of CHD at the first screening level does not exceed 30%. Since the conditions and organization of prenatal examination differ significantly in different countries, this indicator varies very widely from 5% to 63% [4]. It should be emphasized that the best indicators of prenatal diagnosis of CHD are demonstrated by countries with a high level of organization of prenatal diagnosis services [5,6]. The introduction into clinical practice of a qualitative assessment of a four-chamber section of the fetal heart and sections through the main arteries allowed to increase the prenatal diagnosis of CHD by more than 2 times – from 23 to 52% [5]. However, prenatal ultrasound diagnosis of breast cancer is particularly difficult, since the researcher, when evaluating a four-chamber section, needs to achieve a clear image of the partitions throughout, and this is not possible in all cases. Difficulties in diagnosing intracardial septum defects may be related to the peculiarities of fetal position during the study. Thus, the thin atrial septum or the membranous part of the interventricular septum is quite often poorly visualized when using an average class of ultrasound devices or is not visible at all when the cross section of the spine is located "at 6 o'clock". In order to avoid errors in the prenatal diagnosis of intracardial septum defects, it is necessary to detect the defect in at least two planes, one of which must pass at right angles to the septum. Echocardiographic diagnosis of extensive breast cancer is based on the identification of an echonegative zone in its structure, indicating a lack of septum integrity. The greatest difficulties are associated with the diagnosis of FGM, localized mainly in the muscular part of the septum and the infundibular part of the right ventricle. Only a thorough examination of the four-chamber section of the fetal heart makes it possible to identify these defects primarily due to the asynchronous movement of the interventricular septum. Color Doppler mapping provides significant assistance in the prenatal diagnosis of breast cancer. In fetuses with isolated DMF, color Doppler mapping clearly shows significant systolic bypass of blood from the right ventricle to the left and a short diastolic left-right shunt. With extensive large VHD, a monochromatic color signal is more often visualized, and with small ones, a mosaic signal indicating the turbulent nature of blood flow [4,6]. A significant part of breast cancer, including large ones, are detected only in the postnatal period. Submitted by M. Rustico et.al. [4], in 31 (73.8%) of 42 undiagnosed CHD, they were represented by intracardial septum defects. Similar data are provided by E. Tegnander et.al. [5] – 57 (85,1%) out of 67. According to the results of S. Stoll et.al [7], the sensitivity of prenatal echography in the diagnosis of FGM is only 6.6%. According to S. Levi et.al [6], only 8 (13.3%) of the 60 intracardial septum defects were detected during the screening study. Even with a targeted examination at the prenatal diagnostic center, no more than 20% of all isolated DMPP and DMZHP are detected [6]. At the same time, in some specialized prenatal diagnostic centers, the accuracy of diagnosis of FGM reaches 43% [5]. The results of the research obtained by T. Todros et. al. [5,6], showed that even large FGM can be missed by careful examination of the four-chamber section of the fetal heart. In their studies,

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among 11 undiagnosed CHD, 4 cases showed extensive breast cancer. According to the multicenter analysis carried out in 12 European countries, the accuracy of prenatal diagnosis of isolated intracardial septum defects in the late 90s was only 7-8% [7]. When intracardial defects were combined with extracardial anomalies, the accuracy of their prenatal diagnosis was significantly higher and amounted to 40.2 and 31.8%, respectively [7]. In studies conducted by M.V. Medvedev in 1995-2000, it was found that in the conditions of the center for prenatal diagnosis, only 28.3% of breast cancer can be diagnosed, and when small, clinically insignificant defects are excluded from the analysis, the sensitivity of prenatal echocardiography in relation to this type of cardiac anomaly increased to 85% [5]. S.G. provides similar data. Ionova [3], according to which in 2001-2002, 32.5% of septal defects (14 out of 43) were detected at the Prenatal Diagnostic Center in Orenburg.

The purpose of the study

The aim is to improve the ultrasound diagnosis of breast cancer using transvaginal echocardiography in early pregnancy.

Materials and methods

Various congenital heart defects were reported in 182 fetuses/newborns, among which breast cancer accounted for 25.8% (+3.2) or 47 cases. The age of the patients in the study group ranged from 18 to 40 years old. It should be noted that the main group consisted of patients of middle childbearing age (95.7% +3.0) and only two (4.3%+3.0) patients belonged to the older age group. Ultrasound examinations were performed on LOGIC 700 pro serias, VOLUSON 730 PRO and VOLUSON 730 EXPERT devices using B–mode, CDK mode and pulse Dopplerography. In addition, 3/4D modes for fetal heart examination were used for early prenatal diagnosis: DiagnoSTIC technology, TUI, inversion, glass body. Cytogenetic studies were performed in the clinical laboratory of maternity hospital No. 5 and the regional diagnostic center for medical genetics. The pathoanatomical verification of the prenatal diagnosis in cases of medical termination of pregnancy in the second trimester was carried out in the Krasnoyarsk Regional Pathology Bureau with the participation of ultrasound diagnostics doctors of our department, a geneticist.

Results and discussion

The results of prenatal diagnosis of the studied nosological form of CHD are presented as follows: early prenatal diagnosis was possible in 11 (23.4%+6.2) fetuses from 11 weeks of one day to 13 weeks of six days of pregnancy, on average 12 weeks six days and was carried out according to generally accepted diagnostic criteria. In the second half of pregnancy, ventricular septal defects were additionally diagnosed in 7 (14.9%+5.2) fetuses. In the group of newborns with undiagnosed prenatal CHD, 29 (61.7%+7.1) cases of FGM were registered. Various echographic markers and their combination were registered in 10 out of 11 (90.9%+8.7) fetuses with breast cancer diagnosed in early pregnancy (Table 2). In the group of fetuses with breast cancer diagnosed in the second half of pregnancy with ultrasound examination in the first trimester of pregnancy, 1 (14.3%+13.2) fetus The expansion of the collar space to 2.6 mm and the umbilical cord cyst were noted. Considering the presence of two echographic markers, the patient was offered an invasive diagnostic procedure (aspiration of chorionic villi) in order to study the karyotype of the fetus.

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During cytogenetic analysis, the karyotype of the fetus was 46, XX, without structural and numerical rearrangements. The pregnancy was prolonged and an ultrasound examination at 23 weeks and three days revealed breast cancer in the fetus. The pregnancy ended with an urgent delivery of a female fetus weighing 3,400 grams. At the age of the first month of life, the prenatal diagnosis of breast cancer was confirmed and an additional chord of the left ventricle of the heart was additionally detected during echocardiography. By the year of life, the child's condition is satisfactory, he is registered with a cardiologist at a dispensary, does not need surgical correction. In the group of fetuses with breast cancer that were not diagnosed prenatally during ultrasound examination in the first trimester of pregnancy, 7 out of 29 (24.1%+7.9) fetuses had a "deficit" of coccygeal- parietal size and episodes of bradycardia in the fetus. During ultrasound examination in the second trimester of pregnancy, echographic markers were also noted in these fetal groups, such as cysts of the vascular plexuses of the lateral ventricles of the brain, hyperechoic focus in the left ventricle of the heart, fetal intrauterine development delay and abnormal amount of amniotic fluid (Table 3). A careful study of the ultrasound anatomy of the fetus made it possible to detect various early stages extracardial abnormalities in 6 out of 11 (54.5%+15.0) fetuses (Table 4). In the group of fetuses with breast cancer that were not diagnosed prenatally, there was one case (14.3%+6.5) of fetal extracardial pathology – bilateral pyelectasia, detected in the fetus during an anatomy study at 22 weeks and six days of pregnancy. In the group of fetuses with breast cancer diagnosed in the second half of pregnancy, there were no cases of extracardial pathology. Various chromosomal syndromes were registered in 7 out of 47 (14.9%+10.7) fetuses with FGM (Table 5). It should be noted that chromosomal pathology was diagnosed in 5 (71.4%+17.1) fetuses with DMF already at the end of the first trimester of pregnancy. According to one observation (14.3%+13.2), Down syndrome in a fetus/newborn with ventricular septal defects was registered at 22 weeks of one day of pregnancy and postnatally. The outcome of pregnancy in the study group of fetuses was more often noted as favorable (Table 6). Pregnancy ended with urgent delivery and the birth of live children in 37 (78.8%+6.0) patients. Surgical correction of the defect was required for three out of 37 newborns (8.1%+4.5). All 37 children are registered with a pediatric cardiologist at the dispensary, as they require specialized supervision. An unfavorable perinatal outcome was noted in 10 (21.2%+6.0) cases and was primarily due to a frequent combination of congenital heart disease with extracardial and chromosomal abnormalities. A case of neonatal mortality in one (2.1%+2.1) full-term newborn occurred after surgical correction of a defect of the supraortic defect of the interventricular septum with circulatory disorders of stage II "b" and pulmonary hypertension at 6 months of life. Thus, with the help of transvaginal echocardiography performed in pregnant women with echographic markers and congenital malformations, DMF can be diagnosed in 23.4% of fetuses already in the early stages of pregnancy. It is well known that FGM can often occur in fetuses with various chromosomal and genetic syndromes, which can be accompanied by mental retardation and high mortality in infancy and childhood. Therefore, a prenatal examination for the detection of FGM should necessarily include the determination of the karyotype and a detailed study of the ultrasound anatomy of the fetus. Special attention should also be paid to collecting a family history to exclude possible inheritance of CHD. Therefore, there is no doubt that it is necessary to establish a prenatal diagnosis at an earlier stage of pregnancy, since this allows patients to avoid most of the psychological problems that inevitably arise when a gross pathology is detected in the second trimester, incompatible with life and requiring termination of pregnancy for medical reasons.

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Conclusion

Ultrasound examination of the fetus is an important part of pregnancy management. It depends on the results obtained whether the pregnant woman will undergo additional examinations (karyotyping), terminate or carry out this pregnancy.

Obstetricians and gynecologists should prescribe screening diagnostics for all pregnant women within 20 weeks of pregnancy, with an emphasis on ultrasound examination of the fetal heart. The difficulties of the examination (pronounced subcutaneous tissue of a woman, polyhydramnios, lack of water) are indications for repeated examination at 24-25 weeks of gestation.

For the timely detection of CHD, prenatal diagnostic doctors need to improve fetal heart examination skills. The largest number of detected cases in the period after the second ultrasound screening indicates that the women were examined insufficiently during the period of 18-22 weeks of pregnancy.

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