

Ultrasonography in prenatal screening

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Abstract

Ultrasonography is a technique for imaging internal organs with using acoustic waves – ultrasound. It is applied in prenatal diagnostics – this is the second most frequent case of ultrasound examinations. They provide the most valuable information among all methods used in prenatal diagnostic. Performing an ultrasonography with a vaginal probe allows you to recognize the first perceptible symptoms suggesting pregnancy. Ultrasonographic examination allows non-invasive assessment of the embryo, placenta, correct pregnancy, early diagnosis of possible developmental defects in the child, and determination of sex and multiple pregnancy. Thanks to them, it is also possible to recognize the risk of pregnancy threatened. Even before the onset of the first symptoms. Among the techniques of performing prenatal ultrasonography, in addition to classic 2D ultrasonography, three-dimensional ultrasonographies methods are distinguished, as well as Doppler blood flow testing. Each of them provides information on a different diagnostic value in the assessment of regularity and disorders during pregnancy. The popularity of the use of ultrasonography in this area is not only due to its diagnostic value but also because of the safety of its use in pregnant women for both mother and child.

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Ultrasonography

Ultrasonography is a technique of imaging on devices using acoustic waves – ultrasound. The examination was carried out using special apparatus that uses ultrasonic phenomena. Ultrasonic waves sent into the body are reflected from the examined tissues. The phenomenon of acoustic wave reflection on the border of services and various computing

services, sizes and structures on internal devices. It differentiates between solid features and fluid features [1].

Ultrasonography examination has no harmful effect on the human body. The machinery emits an ultrasound beam for about 1/100 of the time it is received. When it show frozen picture the probe does not emit ultrasound. The services offered by ultrasound far outweigh the risks [2].

Pregnancy ultrasonography

Prenatal ultrasonography is the second most common ultrasound examination. At the beginning of pregnancy, the vaginal probe gives the best picture, while from the 4th month of pregnancy, transabdominal probes are more important. The examination provides information on whether the pregnancy is single or multiple and whether it is correctly positioned. The Polish Gynecological Society recommends that at least three ultrasound examinations be performed during pregnancy. The first should be done around 10 weeks of pregnancy, the next about 20 weeks of pregnancy, and the last after 30 weeks of pregnancy. This examination are performed to assess the current development and condition of the fetus. In some cases, it is possible to recognize early the risk of pregnancy a few days before the onset of problems [3].

Ultrasound examination is one of the most valuable tests in prenatal diagnosis of malformations. It is recognized that ultrasound performed in the second trimester of pregnancy allows diagnosing about 80% of major malformations. These defects include: hydrocephalus, microcephaly, absence of skull bones, headlessness, cleft spine, cleft lip and palate, kidney agenesis, diaphragmatic hernia, coating defects, fluid in the abdomen, bone diseases, bone dysplasia, achondrodysplasia [4].

Ultrasonography is performed to:

1. Determination of pregnancy age and fetal weight;
2. Assessment of the rate of intrauterine development and fetal growth;
3. Fetal heart rate analyzes from 6 to 7 weeks of pregnancy;
4. Amniotic fluid volume assessment (oligohydramnios, polyhydramnios);
5. Control the amniocentesis process;
6. Child sex recognition;
7. Determination of placental maturity and placement, exclusion of abnormalities (placenta previa, placental abruption) [5].

Today, ultrasound plays a huge role in recognizing pregnancy and controlling its early development. In

the first trimester of pregnancy, the most practical is an ultrasound examination made by vaginal probe. Thickening of the endometrium is the first visible symptom in ultrasonography suggesting pregnancy, but it does not always confirm its presence. With endometrial thickening, the endometrium enlarges.

The earliest occurring ultrasonography symptom for observing pregnancy is the presence of a gestational sac in the uterus – a hypoechoic formation surrounded by a ring with increased echogenicity [6].

Ultrasonography in the first trimester of pregnancy

The vaginal probe ultrasonographic examination in the 5th-10th week of pregnancy is an important element of early diagnostics, thanks to which it is possible to assess the development and course of pregnancy. According to the current standards of the USG Section of the Polish Gynaecological Society, the first examination must be performed up to the 10th week of pregnancy. It determines the following elements:

- Location of the fetus;
- Pregnancy age – based on the foetus's parietal length and the diameter of the gestational sac;
- Presence, size and location of the gestational sac;
- The number of embryos, choruses and amniotic diseases;
- Presence of heart function;
- Assessment of genital organs – uterus and cervix [7].

The ultrasound examination in the early first trimester of pregnancy can be performed using both the transabdominal probe and the transvaginal probe. Examination by means of transvaginal (endovaginal) technique is more accurate and more common. Before the examination, the woman is recommended to empty her bladder. In the case of examination through the abdominal wall, bladder filling is a necessary condition for such an examination.

The first element of the ultrasonographic examination in the initial pregnancy is the visibility of the corpus uteri. The organ is presented in the longitudinal

plane in the sagittal section, at the same time the anterior-posterior dimension and the width of the corpus uteri are assessed. The correct volume of the uterus in a non-bearing woman is about 100 cm³ and usually does not exceed 125 cm³ in a whale. The next stage of the examination is imaging of the gestational sac, yolk sac, embryo or embryos, observation of embryo movements and heart function. The presence of both ovaries must then be confirmed [4].

The ability to show the gestational sac on an ultrasound depends on many factors, which include: the quality of the ultrasound apparatus, the type of probe used (endovaginal or transabdominal), the skills and experience of the person performing the examination, the position of the uterus (overlap, backbone). The follicle should be visible by the end of the 5th week of pregnancy at the latest when the concentration of β -hCG (human chorionic gonadotropin) is 1000 – 1500 mIU. The visibility of the gestational sac is the first sign of pregnancy on ultrasonography. The standard image of the gestational sac is a hypoechogenic regular fluid area surrounded by hyperechoic fluid. The follicle can be observed around the 4th week of pregnancy and its growth is 1 mm diameter per day. This dynamics persists until the 9th week of pregnancy. In normal pregnancy the follicle grows at the rate of 1 – 1.2 mm per day and reaches the diameter of 5 mm on the 36th and 37th day of pregnancy. The first measurement to determine the gestational age is the size of the gestational sac [8, 9, 6].

In the case of ectopic pregnancy, a similar structure has been proven to exist within the uterine cavity. In this situation, the term “pseudoventricle” is used. Usually, there is no typical hyperechoic echo around it and its shape is irregular and elongated [9].

In a developing pregnancy, another element indicating its growth is the appearance of a yolk sac. The image shows a regular, rounded area of fluid surrounded by hyperechoic echo. It appears around the 5th week of pregnancy. The diagnosis of the yolk sac in the gestational sac is an important symptom confirming intrauterine pregnancy and at the same time excluding the occurrence of germ-free and ectopic pregnancy [8].

When the diameter of the yolk sac exceeds 7 mm this usually suggests abnormal development of early

pregnancy. Abnormal development of the fetus is also indicated by the situation when during the endovaginal transducer examination the yolk sac cannot be visible with the simultaneous occurrence of a pregnancy follicle larger than 12 mm. The confirmation of the presence of fluid of increased echogenicity in the yolk sac, irregular shape or tendency to move within the gestational sac also indicates poor prognosis of pregnancy. The absence of the yolk sac in ultrasound examination at the size of the gestational sac exceeding 20 mm indicates pregnancy pathology [9,6].

Vaginal ultrasound in the first trimester of pregnancy also evaluates the chorion: its size, location, echogenicity and possible irregularities. During the examination, blood extravasations can be observed, their surface, location and attitude to the umbilical cord trailer can be determined. The assessment of the chorion in the first trimester of pregnancy is important because the source of bleeding from the reproductive tract can be identified [10]. During the endovaginal probe examination, an image of the embryo can be presented during the 6th week of pregnancy. Starting from the 6th week of pregnancy the embryo grows about 1 mm per day. This allows you to correctly assess the size of the pregnancy. The ultrasound-measured embryo length of 5 mm is equal to the length of pregnancy lasting 6 weeks and 5 days [11].

Above the 7th week of pregnancy, you can measure the length of the embryo using the CRL (Crown – Rump – Length – parietal – sedentary dimension). The measurement is correctly performed in the long axis, with the fetus completely straightened out and the maximum magnification of the apparatus applied. Accurate determination of the gestational age based on this method is more difficult in advanced pregnancy [6]. The fetus dimensions (biometrics) can be assessed during the second and third trimester of pregnancy using the following measurements: head circumference, abdominal circumference, bipartite dimension and femoral length. The majority of modern ultrasonographic machinery devices automatically calculate a similar fetus mass and the so-called biometric gestational age.

The first movements of the fetus can be seen between the fifth and sixth week of pregnancy. In an

ultrasonographic examination performed in the 7th-8th week of a properly developing pregnancy, individual structures of the fetus can be seen, such as: head, trunk and limbs [12].

The ultrasound examination performed in the first trimester of pregnancy is also the best method of early diagnosis of multifetal pregnancy. It gives the opportunity to determine whether the pregnancy is two – or one – hormone and two – or one – hydrated [13].

In addition to the above mentioned ultrasound examination in the first trimester gives an opportunity to assess the risk of the most common chromosome aberrations (trisomy 21,18,13) using the following ultrasound markers: fetal heart rate (FHR) and fetal neck translucency (NT, nuchal translucency). Moreover, additional ultrasound markers of chromosome aberrations are analysed: fetal nasal bone (NB, nasal bone), tricuspid regurgitation (TR, tricuspid regurgitation), and venous duct pulse index (DV PIV) [14].

Ultrasonographic examination between the 11th and 14th week of pregnancy allows to increase screening activity for trisomy 21 and other trisomies. The examination carried out during this period gives the possibility to determine the duration of pregnancy and is also important in diagnosing many clinically significant defects of the fetus. It allows for the diagnosis of twin pregnancy with the determination of its cosmosity, as well as screening for pre-eclampsia.

In order to determine the risk of chromosome aberrations between the 11th and 14th week of pregnancy, the so-called ultrasound markers are looked for. These are abnormalities which are not considered congenital defects, their commonest cause being disturbances in the development of specific tissues or organs [15].

The symptoms of trisomy of the 21st pair of chromosomes that appear on ultrasound examination are: lateral ventricular dilatation, atrioventricular septal defects, shortening of long bones, thickening of the neck fold (> 3 mm in the first trimester and > 6 mm in the second trimester of pregnancy). In the trisomy of 18 chromosome pairs (Edwards syndrome), ultrasound examination enables the recognition of cardiac defects as well as other features characteristic of this syndrome, such as fetal hypotrophy, shortened

limbs, small head of a characteristic shape and cleft palate [11].

In many cases, it is possible to show the heart's function before the exact visibility of the embryo by using the endovaginal head. During the 5th week of pregnancy, the fetal echo of 2-4 mm in length together with heart function (FHR) can be recorded using the vaginal probe ultrasound. The reason is the rhythmic motion of these muscles visible along the edge of the yolk sac. About the fifth week of pregnancy, the heart rate reaches a low value, between 60 and 90 beats per minute. This activity increases rapidly between 6-10 weeks; the maximum values, i.e. 180 beats per minute, are obtained around the 10th week of pregnancy [13,9,16]. The finding of intrauterine fetal life is one of the symptoms of proper fetal egg development. The vast majority of pregnancies with visible fetal heart function develop properly [11].

Sometimes it is difficult to observe the fetus heart rate, even when the embryo is already 4 mm long. When the embryo is much longer than 4 mm and its heart rate is still invisible, this is a symptom indicating abnormal development of early pregnancy. The diagnosis of fetal bradycardia suggests an increased risk of pregnancy loss. The majority of authors describe that persistence of heart function below 100 beats per minute in the 6th and 7th week of pregnancy significantly increases the risk of miscarriage [4].

The best quality images of the fetus heart can be obtained in the 20th to 22nd week of pregnancy. Abnormal ultrasonography results, abnormal ultrasonographic flows, cardiac arrhythmias and pathological findings of neck translucency are indications for foetal echocardiography [17].

USG in the second trimester of pregnancy

In the second trimester of pregnancy ultrasound is performed to diagnose fetal development disorders. According to the recommendations, the examination is best performed between the 18th and 22nd week of pregnancy, the anatomical structure during this period allows for the best imaging of the fetus structures [18].

It is considered that ultrasonography performed in the second trimester of pregnancy allows to diagnose about 80% of large developmental defects. These defects include: hydrocephalus, headlessness, fluid in the abdominal cavity as well as skeletal diseases, bone dysplasia and others [4].

The study performed in the 18 – 22th and 28 – 32th week provides important information about gestational age, fetal size with simultaneous analysis of the growth process, allows to evaluate the fetus and determine the volume parameters of the placenta, amniotic fluid and umbilical cord. Based on the measurement of BDP and femoral length, the gestational age can be determined with an accuracy of 7 – 10 days. In addition to the biometric measurements, the ultrasound examination evaluates individual structures and organs of the fetus. The examination begins with the determination of the number of fetuses, visual examination of the uterus muscle and the area of adnexa in terms of myomas, malformations and tumours, determination of the correct location of viscera. Then, using ultrasound in the second trimester, a detailed assessment of the fetal organs for the presence of congenital defects is performed. Special attention is paid to the fetal growth process towards the diagnosis of fetal hypotrophy and accurate assessment of the heart, abdominal cavity and brain [18].

Doppler examination

The Doppler examination illustrates blood flow in large arteries and veins using changes in ultrasound wavelengths reflected from moving blood cells. In obstetrics, the Doppler test is used to assess the blood flow in the umbilical artery; it enables early diagnosis of some complications threatening the normal development of pregnancy [7].

It is currently one of the most important studies in standard perinatal care. Analysis of fetal vascular flow helps to assess the severity of fetal anaemia, as the fetal heart ejection volume increases analogously to the decrease in hematocrit values. Doppler examination consists in measuring the peak velocity in

the fetal brain's central artery. This velocity increases simultaneously with the development of pregnancy. The measurement allows to examine the severity of hemolytic disease in the fetus. In the case of anaemia, the test shows an increased ejection volume and a decrease in blood viscosity, which cause an increase in blood flow in the fetal brain's central artery. The use of the Doppler method in prenatal diagnostics contributed to the elimination of the need for invasive tests, including amniocentesis and cordocentesis in over 70% of patients [9, 4].

Doppler ultrasound is a non-invasive and well available screening method for detecting functional disorders of the placenta in the early stages of pregnancy, which is used to predict many abnormalities associated with the process of placenta development and implantation [9].

3D ultrasonography

Currently, in prenatal diagnostics, apart from classical 2D ultrasonography, 3D techniques have been applied, which precisely show the process of embryogenesis and enable visualisation of the fetus at the beginning of pregnancy. 3D ultrasonography enables precise assessment of spatial morphology without the assumption of simple geometric models. The operator has access to dynamic anatomical information through interactive display of objects in 3D space. The benefits of 3D ultrasonography include: better quality of data visualization, volumetric measurements are made based on real measurement data, shortened survey time and the possibility of evaluating spatial objects [1].

It is worth noting that the condition for ultrasound using the 3D method is a favourable position of the fetus and an appropriate amount of amniotic fluid. Difficulties appear in the case of a small amount of amniotic fluid, bad fetal position, presence of umbilical cord loops and the position of limbs above the fetus face. In these cases, proper three-dimensional imaging of the fetus is impossible. These obstacles prevent the proper three-dimensional imaging of the fetus [19].

2D and 3D ultrasound comparison in prenatal diagnostics

The literature contains information related to the potential of using 3D technology and its limitation [10].

According to the available data, the effectiveness of NT measurement (fetal neck translucency) with method 2D is 95.6% and with method 3D 90.4%. The quality and repeatability of the results obtained are relatively comparable, however, limitations occurring in the three-dimensional technique, such as: lack of sufficient amniotic fluid or paranasal fluid, unfavourable fetal position or other and difficulties in obtaining the correct section, give the two-dimensional method the first place in performing and imaging measurements of fetal neck translucency.

According to the researchers, 98% of the body of the corpus callosum can be visualized by three-dimensional ultrasonography. The results showed statistically significant discrepancies in the dimensions of structures imaged by 3D and 2D methods. The results obtained with the three-dimensional technique were significantly higher and were characterized by lower dependence on gestational age. Currently, two-dimensional ultrasonography is the most popular method of imaging the fetal central nervous system.

Recent scientific studies have confirmed an increase in the accuracy of palatal cleft recognition by means of three-dimensional ultrasonography. Older publications compare the precision of 2D diagnosis with 3D. In 2D method for cleft palate is 50% and in 3D 90%. The effectiveness of the classical two-dimensional method in diagnosing lip and gingival clefts in the fetus is between 45% and 68%, while in diagnosing cleft palate, the method is less effective. The use of 3D ultrasonography guarantees much greater effectiveness in diagnosing cleft palate, lip and gingival bifurcation in the fetus. Simultaneous use of both imaging techniques gives accurate results and facilitates making an accurate diagnosis [19].

Three-dimensional ultrasound diagnostics is not more effective than standard fetal imaging methods.

Only in the research on accurate imaging and determination of the type of cleft palate has it been possible to demonstrate a significant advantage of 3D ultrasound over 2D. Some of the other research directions require further analysis to clearly determine their usefulness and application in clinical practice. The undeniable advantage of 3D ultrasonography is that it enables better visualization of the fetus to the parents and presentation of selected anomalies. Two-dimensional ultrasonography remains the “gold standard” in obstetric practice, and three-dimensional methods make it possible to extend and improve the prenatal imaging tests performed so far. Numerous studies on the techniques of three-dimensional ultrasonography indicate a further direction of development in this field and in the future may contribute to increase the possibilities and effectiveness of prenatal diagnostics [19].

Summary

Ultrasonography is the basic method used in prenatal diagnostics. It provides information on the correct course of pregnancy and fetal development. It enables early detection of abnormalities and disorders, and then the initiation of treatment procedures adapted to the observed defects.

Despite the development of ultrasonic wave diagnostics technology, classical two-dimensional ultrasonography is the most valuable test. Three-dimensional ultrasound is becoming increasingly popular due to its importance in the diagnosis of cleft palate. In this respect, it achieves better results than classical ultrasound. An additional advantage, especially for parents, is better visualization of the child, because in the two-dimensional technique, parents encounter difficulties in reading the image visible on the monitor. However, in terms of diagnostic value, the two-dimensional method provides more benefits. It should be remembered that the quality and value of ultrasound results depends not only on the type of method but also on the type of equipment and the skills and experience of the person performing the examination.

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