

## PRENATAL DIAGNOSTICS AND MEDICAL GENETICS

### Available services:

1. First trimester combined screening - BASIC
2. First trimester combined screening - ADVANCED
3. Integrated second trimester test
4. Detailed ultrasound morphology at week 20 to 22
5. Special ultrasound scanning
6. Ultrasound scan focused on fetal growth and placental function at week 36
7. Consultation with clinical genetics
8. Invasive procedures (CVS, AMC)
9. Non-invasive prenatal testing (NIPT)
10. 3D/4D ultrasound imaging

### Benefits:

- examination performed by an experienced medical team
- comprehensive prenatal care at one place
- state-of-the-art instrumentation
- pleasant environment in the city centre
- short appointment times
- individual care

### 1. First-trimester combined screening BASIC

This examination is aimed at screening for the most frequent fetal chromosomal and morphological defects in the period between weeks 11 and 14 of pregnancy.

It consists of 2 parts:

1. biochemical tests performed preferably between weeks 9 and 10 of pregnancy (blood samples to be taken from a pregnant woman for testing blood levels of two hormones - free  $\beta$ -HCG and PAPP-A)
2. ultrasound scanning between weeks 11 and 14 of pregnancy

After completing the ultrasound scan, each woman's individual risk is immediately calculated using a special program that the baby will be born with one of the most common chromosomal abnormalities - Down's syndrome (trisomy 21), Edwards syndrome (trisomy 18), and Patau's syndrome (trisomy 13). This individual risk is calculated from the following data:

- age of pregnant woman
- weight of pregnant woman
- data from patient's history (e.g. ethnic origin)
- results of biochemical tests
- ultrasound parameters (nuchal translucency, nasal bone, etc.)

The pregnant woman will be informed of the next proposed procedure according to the individual risk calculated as above.

If the result is NEGATIVE, the calculated individual risk of chromosomal defects is low and the pregnant woman is usually advised to have a detailed fetal ultrasound morphology in weeks 20 to 22.

If the result is POSITIVE, the calculated individual risk for one of the chromosomal defects is so high that a pregnant woman is advised to have an invasive examination (sampling of chorionic villi = a part of the placental tissue or amniotic fluid collection) to definitively exclude the defect.

**It should be noted that the POSITIVE result of the test does not mean that the child is affected, and on the contrary, the NEGATIVE result of the test does not guarantee the birth of a healthy and living child.**

As part of the first trimester ultrasound scanning, it is also possible to diagnose up to 50% of morphological (structural) fetal defects (e.g. brain, limb, heart, abdominal wall defects, etc.). However, performing a detailed ultrasound scan between weeks 20 and 22 of pregnancy, when up to 90% of serious morphological defects can be detected, is considered necessary by us.

This examination is not reimbursed from public health insurance, but all health insurance companies offer full or at least partial reimbursement of the costs associated with this examination. For more information, please visit the website of your health insurer.

**PRICE: CZK 1,400**

## **2. First-trimester combined screening ADVANCED**

In addition to screening for the most common fetal chromosomal and morphological defects (see above), this examination is aimed at screening for pregnant women who develop an early preeclampsia (about 2% of pregnant women) or placental insufficiency and consequently fetal growth restriction (about 5% of pregnant women). The examination is supplemented with another analyte in pregnant serum (PIGF - Placental Growth Factor), blood pressure measurement and ultrasound of uterine arteries.

PRE-ECLAMPSIA is a serious disorder caused by pregnancy, which can lead not only to the premature termination of pregnancy and delivery of a very premature newborn, but also to the death of a woman, fetus or newborn in the most serious cases.

FETAL GROWTH RESTRICTION is the condition where impaired placental function is most commonly the cause of delayed fetal growth with potential other consequences.

A positive test result for both of these complications is observed in about 10% of pregnant women, but the disease develops only in part of them. The incidence of these diseases can be effectively reduced by up to 50% by early treatment with acetylsalicylic acid 150 mg daily. Special ultrasound scans at weeks 20, 28, 32 and 36 of pregnancy will allow to diagnose the disease more precisely or potentially plan a iatrogenic delivery considering the actual placental functioning and fetal needs.

**PRICE: CZK 1,700**

## **3. Integrated second-trimester test**

The result of the first-trimester combined test is usually supplemented by triple test at week 16. This is a biochemical test (taking blood from the pregnant to evaluate levels of 3 hormones - free  $\beta$ -HCG, uE3 and MS-AFP). Combining the results from the first-trimester combined test and the triple test is used to more accurately calculate the individual risk of Down's syndrome and the risk of spinal defects.

## **4. Detailed ultrasound morphology at weeks 20 to 22**

Ultrasound scanning, which is performed preferably between weeks 20 to 22 of pregnancy. It is used for a detailed assessment of fetal organs and organ systems (central nervous system, skeleton, heart, lungs, digestive and excretory systems, limbs, genitals, umbilical veins, etc.) It can identify up to 90% of morphological (structural) defects and assess chromosomal markers at the same time. It is not covered by public health insurance (beyond the care indicated by a doctor).

**PRICE: CZK 1,000**

## 5. Special ultrasound scanning

Examination that can be performed at any time during pregnancy outside regular examinations when regular ultrasound scan provides unclear findings or if any congenital defect is suspected in the fetus.

When finding a congenital defect, the next step depends on the nature and severity of the defect. Some cases require a more detailed examination, such as invasive examination, magnetic resonance imaging, or additional ultrasound scans to assess the development of the anomaly. Any other procedure should be consulted with both the pregnant and the partner.

## 6. Ultrasound scan focused on fetal growth and placental function at weeks 35 to 36

During this scan, we use Doppler ultrasound to measure fetal size and placental function in order to detect delays in growth and their possible consequences. It is not covered by public health insurance (beyond the care indicated by a doctor).

**PRICE: CZK 1,000**

## 7. Consultation with clinical genetics

Genetic consultation is intended to express the risk of a disease, or to diagnose a genetic disease in the fetus and propose further procedures. Genetic consultation may be indicated for various reasons:

- positive result of the first-trimester combined screening
- positive result of the second-trimester integrated test
- finding of a potential anomaly during the first- or second-trimester ultrasound
- occurrence of a congenital defect or hereditary disease in the pregnant woman's or partner's family

## 8. Invasive procedures

The following invasive procedures are commonly performed: **chorionic villus sampling (CVS)** and **amniotic fluid test (amniocentesis)**. These are currently the only two methods that can definitively exclude chromosomal defects in the fetus. Both methods are performed on an outpatient basis. In chorionic villi sampling (CVS), a small amount of placental tissue is taken under ultrasound guidance for further examination. This is an outpatient procedure performed between weeks 11 and 14 of pregnancy.

For amniotic fluid test (AFT), a small amount of amniotic fluid is taken under ultrasound guidance for further examination. This is also an outpatient procedure that is performed from week 16 of pregnancy.

Complications of both exercises may include a pregnancy loss (0.5%).

## 9. Non-invasive prenatal testing (NIPT)

A simple, safe and accurate screening method that is available to pregnant women to examine Down's syndrome and other common chromosomal defects in the fetus only from mother's blood from week 10 of pregnancy. In this test, a free extracellular DNA of the fetus is isolated from the blood sample of a pregnant woman. The success rate in capturing chromosomal defects is more than 99%.

This method has benefits and limitations, so each examination is preceded by a consultation with a clinical geneticist, who provides the pregnant woman with a detailed explanation about the purpose of the test, the procedure and the meaning of the positive or negative results. Under standard conditions, the test result is available within 14 days. This examination is not covered by public health insurance.

**PRICE: CZK 9,600**

## 10. 3D/4D ultrasound imaging

Three-dimensional (3D) or four-dimensional (4D = real-time 3D, i.e. including detection of fetal movements) imaging can be offered to expectant mothers optimally between weeks 25 to 30 of pregnancy. This imaging technique offers an unparalleled visual experience that can be archived using standard ultrasound pictures or USB media.

Although the 3D/4D imaging is a very promising and fast-developing technique, in this case it is used for commercial purposes only. This is not a medical exam, so the record from this imaging can not be used as forensic evidence when finding a congenital developmental defect. Ultrasound imaging is performed by a trained and qualified nurse rather than by a physician. This examination is not covered by public health insurance.

**PRICE: CZK 1,000**

**For more details, please contact our centre:**

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