Prenatal screening of congenital anomalies

Searching for pregnant women with a significant risk of a specific pathology that can negatively affect the development of the fetus. Screening methods focus on the early detection of chromosomal aberrations and other morphological or functional abnormalities of the fetus. It also focuses on the detection of diseases that are of high risk for the physiological development of the fetus (infection, diabetes, preeclampsia).

In the case of a positive screening result, a diagnostic examination is indicated, which serves to confirm the diagnosis.

1st Trimester Screening

Comprehensive prenatal examination (**up to the 14th week of pregnancy**)[1] Early detection of the most common morphological and chromosomal birth defects of the fetus. The preferred method is the combined test (biochemical and ultrasound), which unfortunately is not covered by health insurance.

Laboratory examination (up to the 14th week)

Determination of **blood count** and blood group **RhD**,also screen for irregular **antierythrocyte antibodies** and serology for **HIV, HBsAg** and antibody against **syphilis**.

As part of the screening for diabetes in pregnancy, we determine **fasting blood glucose**. In case of elevated values (>5.1 mmol/l), the examination is repeated on another day. Values (5.1-6.9 mmol/l) correspond to **gestational diabetes**, (above 7 mmol/l) we speak of **apparent DM**. Regardless of the type of disease, hyperglycemia values must be strictly corrected and stabilized to values in the physiological range (hyperglycemia is a probable cause of the pathogenic effect on fetal development).

Ultrasound examination (up to the 14th week)

Determination of the **number of fetuses**, in the case of multiples, we also determine the number of placentas (chorionicity) and fetal membranes (amnionicity). We evaluate the vitality of the fetus and measure the crown-coccal distance (**CRL**). We compare the value with the population standard and accordingly determine the **gestational age of the fetus** and **calculate the expected date of delivery**.[2]

Combined test (***patient reimbursement***)

A test with a **high detection** of all birth defects (80%).[3] It takes into account the values of blood markers, data from ultrasound examination and medical history (age, weight, height, risk factors in family and personal history, course of previous pregnancy, etc.). Today's methods make it possible to detect, in addition to the risk of developing chromosomal defects, the risk of developing preeclampsia, fetal growth restriction and premature birth.

In addition, the gestational age of the fetus can be determined, multiple pregnancies can be detected, the anatomy of the fetus, the amount of amniotic fluid and the location of the placenta can be fully evaluated.

1. **Blood collection (11+0 to 11+6)** - determination of PAPP-A, free-βhCG and PlGF levels.
2. **UZ examination (11+0 to 13+6)** - measurement of CRL, specification of gestational age, presence of nasal bone, measurement of NT (cervical clearing), tricuspid regurgitation, flow in the ductus venosus and we measure the flow in both *aa. uterinae*.[3]

> Decreased PAPP-A and at the same time increased free-βhCG indicate a high risk for trisomy 21 (Down syndrome), a decrease in both levels is typical for trisomy 13 and 18 (Patau and Edwards syndrome). A decrease in PAPP-A further indicates the risk of developing preeclampsia and fetal growth restriction, very low values can also signal an impending miscarriage. An increase in NT and the absence of a nasal bone are also indicative of Down's syndrome. Increased values of placental growth factor and flow abnormalities in the uterine arteries indicate an increased risk for the development of preeclampsia and fetal growth restriction.

With a positive screening for chromosomal aberrations in the 1st trimester, we indicate **chorionic villus collection** or **amniocentesis**. If the risk of preeclampsia and growth restriction is positive, we administer ASA once a day as a preventive measure until the 36th week.

Non-invasive prenatal diagnosis (***payment by the patient***)

It can be determined **after the 10th week** of pregnancy. We compile the karyotype from the free non-cellular DNA of the fetus **in maternal plasma**. It allowsscreening of **aneuploidies** (Down's, Edwards', Patau's syndrome), **other numerical aberrations** of chromosomes and **gonosomal deviations**. At the same time, it enables the determination of the sex of the fetus and **RhD**.

A suitable alternative to invasive examinations, with a very **high capture**. The disadvantage is that a positive detection must be confirmed diagnostically by invasive methods. This is a relatively expensive examination that is not covered by the health insurance company.

2nd Trimester Screening

We perform regular ultrasound examinations **at the 20th to 22nd week**[1]. We can offer patients a detailed morphological evaluation, but this is not covered by the insurance company.

Ultrasound examination (20-22 weeks)

We evaluate the number of fetuses, vitality, location of the placenta, amount of amniotic fluid. Biometrics include measurements of biparietal dimension (**BPD**), head circumference (**HC**), abdominal circumference (**AC**) and femur length (**FL**) . From these values, the **approximate fetal weight (EFW)** can be estimated.

Detailed morphological ultrasound examination *(paid by the patient)*

Compared to a classic ultrasound, we perform a detailed examination of all organs. We compare their morphology to gestational age and focus on typical signs of morphological defects. We record the results in the protocol.

> We check the presence of both forearm bones and all fingers on the HK, we also observe the shape of the legs. In the facial part, we evaluate the presence of the nasal bone and both eye sockets. We observe the morphology of the brain (butterfly shape) and compare according to physiological findings of the same gestational age. Next, we carry out a careful **heart check** (correct localization, rotation, 2 ventricles and 2 atria, fully separated by septa. We evaluate the flow of the *ductus venosus* and the flow of the tricuspid valve, determine the heart rate, the presence of murmurs. We check for the presence of large blood vessels and their entry into the heart compartments. We evaluate the shape of the stomach, kidneys and calyces. At this point, it is already possible to determine the gender of the fetus.

oGTT (24-28 weeks)

They undergo all women with a negative detection in the 1st trimester. We do it in the morning, after 8 hours of fasting (3 days before the classic eating habits test). We take blood from a peripheral vein on an empty stomach and determine the current blood glucose level. Subsequently, the woman drinks a solution of 75 g of glucose in 300 ml of water (within 3-5 minutes). Another blood sample is taken after the 60th (Gly < 10.0 mmol/l) and after the 120th minute (Gly < 8.5 mmol/l).[4]If there is no **drop in blood glucose to the original value'** even after 120 minutes, the patient is entrusted to the care of a diabetologist.

🔎 *For more information see Oral Glucose Tolerance Test.*

3rd Trimester Screening

In 10% of pregnancies, the growth of the fetus slows down from the second half (in about 7%), we also find a failure of placental functions, which leads to an insufficient transfer of nutrients and oxygen (hypoxic newborn).

Laboratory examination (28-34 weeks)

Determination of blood count and syphilis serology

Ultrasound examination (30-32 weeks)

We evaluate the number of fruits, their vitality and position. We determine biometrics: by measuring BPD, HC, AC, FL, and then calculating EFW. Examination of morphology of organs. We evaluate the localization of the placenta (consider the distance of the lower pole from the inner gate) and the amount of amniotic fluid.

Vaginorectal detection of GBS (35-37 weeks)

Streptococcus agalactiae (type B) is found in 30% of women as a natural part of the vaginal microflora. However, it is the **most common life-threatening disease of newborns** (mortality 20-30%). Infection of the newborn occurs during passage through the birth canal. Risk factors include the delivery of premature newborns, premature outflow of amniotic fluid, low gestational age, fever during childbirth, etc. Early infections (80%) occur under the guise of *neonatal sepsis*. Late ones are more often manifested as meningitis.

In the event of a positive detection, we indicate **ATB of the diaphragm during childbirth** (i.v. penicillin).

Screening of fetal growth restriction (***payment by the patient***)

It is performed in the 36th week. Growth restriction occurs in 5-10% of pregnant women and is the cause of 30-50% of intrauterine deaths. The main goal of this examination is to check the growth of the fetus and its sufficient vascular supply.

We will carry out *biometrics* of the fetus (determination of size and weight). We will determine the Doppler flow parameters by measuring the pulsatility index (a. cerebri media, a. umbilicalis, ductus venosus and aa. uterinae). Subsequently, we create a biophysical profile of the fetus. This will give us information about any risks. And based on this, we can plan the next course of action (including early termination of pregnancy).

Other procedures in the 3rd trimester

- **From the 28th week**: possible to perform antepartum **RhD alloimmunization** in Rh^- mothers.
- **36-37. week*: registration of a pregnant woman** *to the maternity hospital*'.
- **From the 38th week**: women are offered the option of inducing labor using the **Hamilton Maneuver**.
- **From the 40th week**: at the doctor's discretion, we perform a **cardiotocographic non-stress test** (consideration of the hemodynamic stability of the fetus at rest).
- **Between 41+0 and 42+0**: steps are taken to **terminate the pregnancy** (preinduction, induction of vaginal delivery).

Links

Related Articles

- Prenatal diagnosis
- Congenital developmental defects
- Indications for chromosomal examination
- Preeclampsia
- Fetal growth restriction

References

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