Screening of congenital developmental defects

Search for pregnant women at significant risk for a specific pathology that may adversely affect fetal development. Screening methods focus on **early detection of chromosomal aberrations** and other **morphological or functional abnormalities** of the fetus. They also focus on the detection of diseases that are high risk for the physiological development of the fetus (infections, diabetes, pre-eclampsia).

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

Revision of the recommended procedure of the CGS ČLS JEP No. 1/2019 Coll. Validity from January 2021

Screening in the 1st trimester

Comprehensive prenatal screening (**up to 14 weeks of gestation**)[1] Early detection of the most common fetal morphological and chromosomal birth defects. The preferred method is the so-called 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'**, screening for irregular **anti-erythrocyte antibodies** and serology for **HIV**, **HBsAg'** and antibodies to **syphilis**.

As part of the screening for diabetes in pregnancy, we determine **fasting glycaemia**. If the values are elevated (>5.1 mmol/l), we repeat the test on another day. Values (5.1-6.9 mmol/l) correspond to **gestational diabetes**, (above 7 mmol/l) we speak of **overt DM**. Regardless of the type of disease, hyperglycaemia values must be strictly corrected and stabilised to values within the physiological range (hyperglycaemia is a likely cause of a pathogenic effect on fetal development).

UZ examination (up to 14 weeks)

Determination of the **fetal count**; in multiple fetuses, we also determine the number of placentas (chorionicity) and amniotic envelopes (amnionicity). Assess fetal vitality and measure the craniocervical distance (**CRL**). We compare the value with the population standard to determine the **gestational age of the fetus** and **calculate the expected date of delivery**.[2]

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

A test with **high detection'** of all fetal birth defects (80%).[3]

It takes into account blood marker values, ultrasound scan data and medical history (age, weight, height, risk factors in family and personal history, course of previous pregnancy, etc.). In addition to the risk of *chromosomal defects*, today's methods allow the risk of *pre-eclampsia*, *fetal growth restriction* and *premature birth* to be determined.

In addition, the gestational age of the fetus can be determined, multiple pregnancies can be detected, fetal anatomy, amniotic fluid quantity and placental localization can be completely evaluated.

1. **Blood collection (11+0 to 11+6)'** - determination of PAPP-A, free-βhCG and PlGF levels.
2. **Ultrasound scan (11+0 to 13+6)** - measure CRL, clarify gestational age, presence of nasal os, measure NT (nuchal translucency), tricuspid regurgitation, flow in ductus venosus and measure flow in both *aa. uterinae*.[3]

> Decrease in PAPP-A and concomitant increase in free-βhCG indicates high risk for trisomy 21 (Down syndrome), and decrease in both levels is typical for trisomies 13 and 18 (Patau and Edwards syndromes). Furthermore, a decrease in PAPP-A indicates a risk for the development of pre-eclampsia and fetal growth restriction; very low levels may also signal an impending miscarriage. An increase in NT and absence of a nasal ossicle are also indicative of Down syndrome. Elevated placental growth factor values and flow abnormalities in the uterine arteries indicate an increased risk for the development of pre-eclampsia and fetal growth restriction.

When screening positive for chromosomal aberrations in the first trimester, we indicate **chorionic villus sampling** or **amniocentesis**. If the risk of pre-eclampsia and growth restriction is positive, we administer **1x daily ASA** as a prophylactic until 36 weeks.

Non-invasive prenatal diagnosis (***patient reimbursement***)

Can be determined **after the 10th week** of pregnancy. Karyotype is constructed from free non-cellular fetal DNA **in maternal plasma**. It allows screening for **aneuploidies** (Down's, Edwards, Patau syndrome), **other numerical aberrations** of chromosomes and **gonosomal abnormalities**. It also allows the determination of fetal sex and **RhD'**.

It is a suitable alternative to invasive testing, with a very **high detection rate**. The disadvantage is that positive detections must be confirmed diagnostically by invasive methods. This is a relatively expensive test that is not covered by insurance.

2nd trimester screening

We perform regular ultrasound examinations '*at 20-22 weeks*'[1]. We can offer patients a detailed morphological evaluation, but this is not covered by insurance.

UZ examination (week 20-22)

We evaluate the number of fetuses, vitality, placental localization, amount of amniotic fluid. Biometry includes measurements of biparietal dimension (**BPD*), head circumference (****HC**), abdominal circumference (**AC*), and femur length (****FL**). From these values, we can estimate the **approximate fetal weight (EFW)**.[2] We then assess the morphology of tissues and organs, looking for anomalies.

Detailed morphological ultrasound examination (paid by the patient)

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

> We check the presence of both bones of the forearm and all fingers on the HK, we also observe the shape of the feet. On the facial part, we evaluate the presence of the nasal bones and both orbits. We observe brain morphology (butterfly shape) and compare according to physiological findings of the same gestational age. We also perform a careful **heart check** (correct localization, rotation, 2 ventricles and 2 atria, fully separated septa. We evaluate the *dutus venosus* flow and tricuspid valve flow, determine the heart rate and the presence of murmurs. Check for the presence of large vessels and their entry into the cardiac compartments. We evaluate the shape of the stomach, kidneys and calyces. At this point, it is possible to determine the sex of the fetus.

oGTT (24th-28th week)

Undergoes all women with a negative first trimester uptake. Performed in the morning, after 8 hours of fasting (3 days before the test classic eating habits). On fasting, we draw blood from a peripheral vein and determine the current glycemic level. The woman then drinks a solution of 75 g glucose in 300 ml water (within 3-5 minutes). Another blood draw is performed after the 60th (Gly < 10.0 mmol/l) and after the 120th minute (Gly < 8.5 mmol/l).[4]If there is no **fall in glycemia to baseline** even after 120 minutes, the patient is referred to the care of a diabetologist.

🔎 *For more information see Oral glucose tolerance test.*

Screening in the 3rd trimester

In 10% of pregnancies there is fetal growth retardation from the second half onwards (about 7%) we also find a failure of placental function, leading to inadequate transfer of nutrients and oxygen (hypoxic newborn).

Laboratory examination (28-34 weeks)

Determination of **blood count'** and serology for **syphilis'**

UZ examination (30th-32nd week)

Assess the number of fetuses, their **vitality and position**. We determine biometry: by measuring **BPD, HC, AC, FL,** and then calculating **EFW**. Examination of **organ morphology**. Assess **placental localization** (consider the distance of the lower pole from the internal gate) and the amount of **amniotic fluid**.

Vaginorectal GBS detection (week 35-37)

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 rate 20-30%). Infection of the newborn occurs during passage through the birth canal. Risk factors include prematurity, premature amniotic fluid, low gestational age, fever during labour, etc. Early infections (80%) present with *neonatal sepsis*. Late ones more often present as meningitis.

In case of positive detection, we indicate **ATB screen at delivery** (i.v. penicillin).

Screening for fetal growth restriction (***patient reimbursement***)

Performed at 36 weeks. Growth restriction occurs in 5-10% of pregnancies and is the cause of 30-50% of intrauterine deaths. The main aim of this examination is to check the growth of the fetus and its adequate vascular supply.

We perform **biometry** of the fetus (determination of size and weight). Determine the Doppler *flow parameters* by measuring the pulsatility index (*a. cerebri media*, *a. umbilicalis*, *ductus venosus* and *aa. uterinae*). We then generate a biophysical profile of the fetus. This will provide us with information on possible risks. A na základě toho můžeme plánovat další postup (včetně předčasného ukončení těhotenství).

Other procedures in the 3rd trimester

- **From week 28**: antepartum **RhD alloimmunization** can be performed in $^{\text{Rh-maternal}}$ mothers.
- **3 weeks**: Registration of a pregnant woman **to the maternity hospital**.
- **From 38 weeks'*: women are offered the option of induction of labour using*** *Hamilton's touch*.
- **From 40 weeks**: at the discretion of the physician, we perform a **cardiotocographic non-stress test** (assessment of the haemodynamic stability of the fetus in a resting state).
- **Between 41+0 and 42+0**: steps are taken to terminate the pregnancy **(preinduction, induction of vaginal labour).**

Links

Related articles

- Prenatal diagnosis
- Congenital malformations
- Indication of chromosomal examination
- Preeclampsia
- Fetal growth restriction

Reference

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