

Pränatale Diagnostik Berlin - Lichtenberg

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Information on first trimester examination during pregnancy

The ultrasound examination in early pregnancy requires both a high-resolution ultrasound machine and an experienced examiner. The basis of the examination in this period is early organ diagnostics. This is supplemented by a risk calculation for fetal trisomies and the calculation of your individual risk for the occurrence of preeclampsia (pregnancy toxicosis). The ultrasound examination is not harmful to the fetus and is usually performed from the abdomen. If visibility is unfavorable, a transvaginal examination may also be necessary. We would like to briefly explain the individual components of the examination in preparation for the informative discussion prior to the examination.

Early organ diagnostics

The gestational age can be assessed by measuring growth. The fetal organs can be visualized for the first time. At such an early stage, a large number of malformations can be detected or largely ruled out. These include severe anomalies of the head and brain, arms and legs, defects of the urinary tract and abdominal wall. Severe and complex heart defects can also be detected by early fetal echocardiography. As a conspicuous neck with an accumulation of fluid in the tissue under the skin can be an important indication of organic malformations, fetal nuchal translucency is always also assessed.

Nuchal translucency measurement and risk calculation for fetal trisomy 21, 18 and 13 at 11+0 to 13+6 weeks of pregnancy



Fetal trisomies, such as trisomy 21 (Down's syndrome), are more common the older the pregnant woman is. However, a trisomy can also occur in young pregnant women. In first trimester at the age of 20, the probability of trisomy 21 is 1:700, at the age of 25 1:616, at the age of 30 1:415, at the age of 35 1:175 and at the age of 40 1:51.

How high your personal risk is can be calculated statistically on the basis of your age in combination with sonographic criteria in the above-mentioned period. The main criterion is the thickness of the so-called nuchal translucency (nuchal fold). A thickened nuchal fold increases the risk. Other criteria, such as the length of the nasal bone, the blood flow in the Ductus venosus and over the right heart valve (tricuspid valve), can be included in the risk calculation if they can be sufficiently assessed by ultrasound. A distinction is made between three risk areas:

Low risk: < 1:1000

Intermediate risk: 1:100 - 1:1000

High risk: > 1:100

The detection rate for a chromosomal disorder is approx. 80% with ultrasound alone. If you want to rule out the above-mentioned or other chromosomal disorders with certainty, this is only possible using an invasive method (extraction of fetal or placental DNA using amniocentesis or placental puncture). NIPT can be an alternative to invasive diagnostics.

NIPT - Non-invasive prenatal test for trisomy 21 / 18 / 13

This test is an excellent screening test that can detect fetal trisomies 21, 18 and 13 with a very high degree of certainty by taking a blood sample from the pregnant woman. Cell-free DNA fragments of placental origin in the mother's blood are analyzed. If a disproportionate amount of DNA from one of the three chromosomes is found, a trisomy must be suspected. The detection rate is high at 99% for trisomy 21 and approx. 95% for the other two trisomies. The rate of false test results is low (at 1 in 1000 tests carried out). If the test result is inconspicuous, trisomy cannot be ruled out, but it is very unlikely. The test can also be carried out on twins. Chromosomal abnormalities other than those mentioned are not detected by NIPT. For this reason, NIPT is not suitable (and is not covered by health insurance) if the fetus has a malformation. In the case of an abnormal NIPT result, human genetic counseling and clarification with a diagnostic puncture (amniocentesis) is recommended. In about 2% of cases after NIPT, no result can be given by the laboratory. In this case, we will discuss the next steps with you, for example repeating the test at a later date. Whether the test makes sense for you will be discussed during the personal consultation and on the basis of the results of the ultrasound examination. You do not have to decide immediately on the day of the ultrasound examination, but can also come back to us after a reflection period for the NIPT blood test. The costs for the NIPT are covered by statutory health insurance and, as a rule, also by private health insurance. In our practice, NIPT is not carried out isolated, but only together with ultrasound diagnostics, as it cannot replace the examination of the fetus's physical development, but can only supplement it. We will inform you of the result by telephone and send you the written laboratory findings.

Preeclampsia risk (Calculation of the individual risk of pregnancy toxicosis)

As part of the first trimester screening, a risk assessment for possible preeclampsia can be carried out. Preeclampsia is characterized by the following symptoms: high blood pressure, edema (water retention) and protein excretion in the urine. Preeclampsia is often associated with placental insufficiency, i.e. the placenta does not "nourish" the fetus sufficiently. This clinical pattern usually occurs in the last trimester of pregnancy, but can also occur earlier. Premature delivery may be necessary due to the progression of the disease.

If there is an increased risk in your preeclampsia screening, current studies show that this risk and the consequences can be reduced by taking ASA 150 mg (acetylsalicylic acid, also known as Aspirin) prophylactically. This ASA intake should be started until 16+0 weeks' gestation and ends at 36 weeks' gestation. Furthermore, if there is an increased risk of preeclampsia during the course of the pregnancy, additional ultrasound examinations to monitor fetal growth and blood flow (Doppler sonography) are recommended.

Information on ultrasound examinations during pregnancy

Thanks to high-end ultrasound equipment and many years of experience in the field of prenatal medicine, we are able to assess by ultrasound examination the child's state of health and detect or rule out fetal abnormalities, including a large number of fetal malformations and diseases. We ask you to take note of the following information before the examination.

The ultrasound examination is an imaging procedure which, according to current knowledge, does not cause any negative effects or damage to the unborn child. This also applies to repeated examinations.

Even with very high quality equipment, the utmost care and experience of the examiner, not all malformations and changes can be detected at every stage of pregnancy. It is possible that existing anomalies are not detected (e.g. a defect in the cardiac septum, a small cleft lip and palate, small defects in the spinal column, finger/toe anomalies). The ability to assess the child may be limited by unfavorable examination conditions. These include, for example, a reduced

Patienten - ID:

Notes on the personal conversation:

Date and place:

Signature of pregnant woman: Signature of doctor:

If you have any questions about the “General Data Protection Regulation”, “Patients' Rights Act” and “Genetic Diagnostics Act”, please contact us directly. The laws can be viewed at the registration desk.