INFORMATION ON THE **FIRST TRIMESTER SCREENING (FTS)**

DEAR PARENTS-TO-BE,

pregnancy is a unique and joyful experience that is often accompanied by many questions. In particular, the main focus is on the health of the unborn child.

We share your excitement about your pregnancy and would like to use this information leaflet to give you with an overview about the options for an early exclusion of malformations and the limitations, advantages and disadvantages of various examination methods.

As most children are born healthy, the first trimester screening (FTS) can be helpful in reducing possible fears and worries at an early stage. If conspicuous findings or an increased risk are detected, the results can help to take appropriate measures. This could include additional tests, personalized care during pregnancy and birth or – if necessary – special care for the child after birth.

The consultation for these tests is carried out in accordance with the legal requirements. You have the right to decide for yourself whether and which tests you wish to undergo and to whom the results may be disclosed. You also have the right not to know, which we naturally respect.

The best way to decide which examination makes sense in your individual situation is to read this information leaflet. Please read the following information carefully and discuss any questions you may have during your consultation. Our team of experienced specialists in prenatal diagnostics and genetics will be happy to assist you.

PRAE NATAL
PRÄNATALMEDIZIN GYNÄKOLOGIE UND GENETIK

FIRST TRIMESTER SCREENING (FTS)

PART 1:
EARLY ULTRASOUND ORGAN SCAN

First of all, we would once again like to emphasize:

Most children are born healthy and most pregnancies proceed without complications! Only 3 to 4 percent of all newborns present some kind of abnormalities, the majority of which have a good prognosis or treatment options.

A frequent focus of parental concern are trisomies, in particular trisomy 21 ("Down syndrome"), as well as trisomy 18 and 13. However, these only account for a small proportion of the possible anomalies. Other genetic and, above all, non-genetic malformations occur far more frequently.

Severe malformations, especially those of the head, brain, spine, limbs, abdominal wall, bladder, kidneys, diaphragm and serious heart defects, can often be detected during the early ultrasound organ scan (also known as "early exclusion of malformations").

Therefore, an ultrasound performed between 12+0 and approximately 14+0 weeks of gestation is the basis for additional examinations.

Using modern, high-resolution ultrasound equipment, a specialized physician systematically examines the organs and organ systems visible at this gestational stage. In addition, soft markers such as nuchal translucency, the nasal bone and the tricuspid valve are assessed as possible signs of an increased risk of trisomies (13, 18 and Down syndrome) as well as other malformations.

For most couples, the early ultrasound organ scan results are usually inconspicuous and can significantly reduce anxiety and uncertainty. It has also been proven to enhance parental bonding to the child.

If the early ultrasound organ scan shows abnormalities, additional diagnostics and genetic counseling should be performed. The aim is to provide extensive, specific and individualized care throughout the rest of the pregnancy. Determining the risk of trisomy 21, 18 and 13 by taking a blood sample (NIPT) is often no longer sufficient in these cases, as a clear diagnosis is only possible with a prenatal diagnostic test.

The informative value and reliability of the ultrasound depend on the examination conditions and the gestational age. Factors such as echo-rich abdominal walls, the position of the uterus or placenta and the position of the baby can make the assessment more difficult and may require the examination to be repeated. The optimum period for a precise assessment is between 12+0 and 14+0 weeks of gestation.

Even under favorable conditions, the early ultrasound organ scan does not offer maximum certainty in this gestational stage. For this reason, we recommend an additional extended ultrasound of the organs between the 21st and 23rd week of gestation in order to rule out congenital diseases and malformations as far as possible.

PART 2:
RISK CALCULATION FOR COMPLICATIONS OF PREGNANCY – RISK OF PRE-ECLAMPSIA

Pre-eclampsia (formerly also known as pregnancy poisoning or gestosis) is one of the most common complications of pregnancy. It poses a considerable risk to the health of the mother and child, especially if it occurs early in the pregnancy. It is one of the main causes of fetal growth restriction and premature births before the 34th week of gestation.

In many cases, an early detection of an increased risk allows preventive measures to be taken to avoid the occurrence of complications. At the same time, it allows the initiation of specialized care and close monitoring of these pregnancies.

The individual risk for pre-eclampsia is assessed as part of the early ultrasound organ scan. A combination of different parameters is taken into account:

PRE-EXISTING RISKS

- Complications in previous pregnancies
- Family history of pre-eclampsia
- Maternal Age under 18 or over 40
- High blood pressure
- Type I diabetes mellitus
- Obesity
- Kidney disease
- Autoimmune diseases

PREGNANCY ASSOCIATED RISKS

- First pregnancy
- Pregnancy after IVF, ICSI or ovum donation

In addition, a doppler ultrasound of the uterine arteries is performed and maternal blood pressure is taken. These examinations are conducted as part of the first trimester screening. The results are available immediately after the examination.

With this method, the risk of early and severe pre-eclampsia can be predicted with an accuracy of 85% for those actually affected. If an increased risk is detected, the risk of developing pre-eclampsia can be reduced by around 80 percent by taking low-dose Aspirin (acetylsalicylic acid) at an early stage.

In these cases, close monitoring of the pregnancy is recommended and the expecting mother should check her blood pressure regularly.

If the risk of pre-eclampsia is within the normal range, the probability of pre-eclampsia occurring is less than five percent (false negative rate).

Through early risk assessment and appropriate measures, we can contribute to making the pregnancy safer and minimizing risks for mother and child.

NON-INVASIVE PRENATAL TEST (NIPT)

RISK CALCULATION FOR CHROMOSOMAL DISEASES USING MATERNAL BLOOD CONTAINING CELL-FREE DNA (CFF-DNA)

The risk of a developmental disorder in a child caused by chromosomal anomalies is between 0.5 and 2 percent, depending on the maternal age. One of the most common chromosomal anomalies is trisomy 21 (Down syndrome).

For many women, it is important to know their individual risk in order to make informed decisions about possible prenatal genetic testing (e.g. amniocentesis). NIPT can reliably determine the risk for the three most common trisomies (21, 18 and 13) and therefore offers additional safety and guidance.

The early ultrasound organ scan is an important foundation for the NIPT. Generally, this test should only be performed if the ultrasound shows no abnormalities. In certain circumstances, the test can also be considered if there are special findings, but this should be discussed on an individual basis.

NIPT is performed using a simple blood sample from the mother, which is used to obtain and analyze fragments of genetic material from the placenta (cff-DNA). In most cases, this DNA represents the genetic material of the child. The test result provides a probability for the presence of certain chromosomal anomalies (trisomy 21, 18 and 13), but is not a definitive diagnosis.

PROCEDURE AND ACCURACY

Timing

Technically, NIPT can be performed at or after 9+5 weeks of gestation. However, the early ultrasound organ scan is strongly recommended prior to the test and therefore, we recommend waiting until 12+5 weeks of gestation.

Duration

Results are usually available by 7–10 business days.

Detection rates

Trisomy 21: 99 %
Trisomy 18 and 13: approx. 97 %
False-positive-rate (FPR): less than 1 %

An increased risk does not automatically mean that the child is actually affected. It solely indicates that further clarification through genetic counseling and prenatal diagnostic testing (e.g. puncture) is advisable.

Sometimes the results of the test cannot be interpreted, e.g. if the concentration of cff-DNA in the mother's blood is too low. In such cases or in the event of other irregularities, genetic counseling should also take place in order to discuss further steps.

ARE THERE ANY RISKS INVOLVED IN THE FIRST TRIMESTER SCREENING?

The test as such has no physical risks. There are no known complications when modern ultrasound equipment is used by experienced prenatal diagnosticians.

Possible risks are rather associated with the psychological burden that can arise from abnormal test results. In such cases, rapid diagnostics and individual, thorough consultation are strongly recommended. In addition, seeking psychosocial counseling can provide valuable support.

FURTHER PROCEEDINGS

This information leaflet is designed to help you prepare for your first trimester screening in our practice.

In preparation for the examination, we will obtain some information about previous illnesses and risks and take your blood pressure. So please be on time for your appointment!

You will then have the opportunity to discuss any remaining questions with our prenatal diagnostician before the examination. By signing the respective form, you will confirm that you have read and understand this information leaflet.

We look forward to your visit and wish you an uncomplicated and smooth pregnancy.

YOUR PRAENATAL-TEAM

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