Why is this test offered?

■ Risk of fetal trisomies (chromosome errors)

Most children will be born healthy. Still, the chance that chromosome errors arise, which lead to physical and/or intellectual impairment is approx. 0,5-1%.
The most frequent type of chromosome errors is the **trisomy 21, also called "Down-Syndrome"**. The risk for this disease rises with maternal age, thus it is generally recommended that pregnant woman older than 35 should consider an amniocentesis.
However, since younger women have more pregnancies, 70% of children with Down-Syndrome are born to woman younger than 35.

Thus, many younger women wish to know their **individual risk** for such trisomies and based on this information decide on further genetic testing, such as amniocentesis or NIPT.

■ Risk of pregnancy associated complications

Preeclampsia, also known as gestosis is one of the most frequent and also most dangerous complications in a pregnancy.
Particularly early onset preeclampsia has a risk of fetal malnutrition, preterm delivery, stillbirth and even maternal death.

If an elevated risk for preeclampsia is identified, **prophylactic measures** can be initiated to avoid severe complication.

Surname:

Name:

PC-Number:

Date:

■ Information on Aneuploidy Risk Calculation (First Trimester Screening)

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■ Dear Patient,

You've opted for a first trimester screening, comprising an early ultrasound organ scan and personalized risk calculation for trisomies.

This test has evolved since the early 90's from the simple nuchal fold measurement. Today, the first trimester screening comprises testing of numerous parameters from the maternal blood and high resolution ultrasound.
These measurements finally allow a personalized risk calculation **for fetal trisomies** and for a maternal gestosis (preeclampsia).
Furthermore, **many fetal malformations** may be detected or excluded at an early stage.

A normal result will dispel parental fears. On the other hand, if specific risks or abnormalities become apparent further tests or prophylactic measures may be discussed.

The test is subject to the German law on genetic testing (Gendiagnostikgesetz, GenDG). Thus, it is up to your personal discretion which genetic features of the fetus shall be investigated and who shall be informed of the results.

In case of pathological findings in the fetus you are entitled to receive psychological support. This may also be appropriate if i.e. the waiting time for laboratory results should pose emotional stress for you.

As a prerequisite for the test we need your written and signed consent. Please read this information leaflet carefully and ask the genetic counselor or ultrasound doctor if any questions arise.

■ Risk of fetal malformations and/or diseases

For most pregnant woman and their partners an early (13./14. week of gestation) exclusion of fetal malformations will lead **to relief fears** on the development of the baby. This is known to enhance parental bonding to the child.

Still, 3-5% of all newborns present some kind of physical or mental impairment. A large part of these may be reliably detected or excluded in the ultrasound organ scan in the 20th-23rd week of gestation.

Some **particularly severe malformations** may even be detected in the early ultrasound scan, which is part of the first trimester screening. Examples of malformations include severe heart malformations, defects of the abdominal wall or the diaphragm and severe forms of spina bifida.

An early detection allows targeted counseling, planning of the birth management and preparation of therapeutic actions.

How is the test done?

The first trimester screening can be performed between 11+5 and 14+0 week of gestation. It comprises three parts from which the individual risk for a fetal Trisomy 13, 18 and 21 and a maternal preeclampsia is calculated.

1. The concentration of **three placenta derived hormones** will be measured in a maternal blood sample (free ß-HCG, PAPP-A and PlGF).
Low levels of PlGF and PAPP-A point to placental dysfunction and/or possible later gestosis. High ß-HCG and low PAPP-A are indicative for a Trisomy 21 (Down-Syndrome). In pregnancies with Trisomy 13, 18 or a Triploidiy both hormones (PAPP-A and ßHCG) are low.

2. **Taking your medical history will contribute to the risk evaluation.**

Important risk factors are:
- Complications in preceeding pregnancies
- Positive family history for preeclampsia
- Age <18 or >40 years
- High blood pressure
- Diabetes mellitus type I
- Adipositas
- Impaired renal function
- Autoimmune diseases
- First pregnancy
- Pregnancy after IVF / ICSI / ovum donation

3. An experienced investigator will perform a high-resolution ultrasound scan and will measure:
- **Thickness of the nuchal fold**, which is a deposit of tissue water, present in the fetal neck
- Thickness of the fetal nasal bone
- Perfusion of the blood vessels leading to an from the fetal heart
- Fetal heart rate
- **Perfusion of the blood vessel leading from the placentato the fetus**

All these parameters contribute to the risk calculation for trisomies and preeclampsia. In addition, the high resolution ultrasound allows the early detection of many (however not all) fetal malformations.
The combined results as well as any resulting recommendations will be discussed with you directly after the ultrasound scan. Your local gynecologist will receive a written report on all results. If genetic defects such as trisomies are suspected, a genetic counseling session will be offered.

How reliable are the results?

It is important to keep in mind that the tests discussed here are statistical risk calculations but not diagnostic tests. Thus, any positive result does not necessarily imply a disease or pathological situation but merely suggests the consideration of further follow up investigations (like amniocentesis) or preventive measures.
Although the early ultrasound is a sensitive tool to detect a wide range of fetal malformations, even under best conditions it is impossible to detect all aberration or malformations in the fetus.
Thus, we recommend a second organ ultrasound scan in gestational week 20-23, which is particularly suited for the detection of heart malformations.

■ Risk calculation of chromosome errors

The sensitivity of the risk calculation programs depends on the type and number of parameters included.
If just the maternal age is considered, the sensitivity would only be 30%. If the nuchal fold is added, sensitivity rises to 75%. Inclusion of the maternal hormone levels leads to a detection rate of 90% and taking into account **further sonographic marker** such as the nasal bone sensitivity **is as high as 93%.**
In multiple gestations the sensitivity is generally about 10% lower.
Obviously, the aforementioned detection rates require that if an elevated risk is calculated, an invasive diagnostic (amniocentesis) follows.
However, more than 80% of all pregnant woman receive results in the low-risk distribution (risk below 1:1000). Regarding the procedure associated risk for miscarriages of 0,3-1% (1:100-1:300) an invasive procedure for these woman is not recommended.

■ Risk for preeclampsia

The above described procedure for prediction of preeclampsia reaches a detection rate of 85%.
If an increased risk is calculated, a medication **with low dose aspirin (100 mg/d) will significantly reduce this risk and/or diminish the severity of the symptoms. The pregnancy should then be closely monitored for any signs of gestosis**. With normal screening parameters the risk for preeclampsia is below 5%.

What kind of problems might arise in course of the test?

Several factors might impede the first trimester screening.
The ultrasound is sensitive to maternal adipositas. An unfavorable position or flexion of the uterus or position of the fetus might complicate the ultrasound. Best timepoint is between gestational week 12+5 and 14+0. Too early or too late ultrasound often gives unsatisfactory results.
Blood samples for the biochemical parameters (maternal levels for PAPP-A, ßHCG and PlGF) should be drawn between 9+5 and 11+5 gestational week. Blood values might be distorted by heat or long transport duration.
Due to stringent quality control measures in our laboratory erroneous measurement are highly unlikely.

Are there any risks associated with the first trimester screening?

The test as such (ultrasound and drawing blood from the mother) has no physical risks.
However, waiting for the result or receiving a suspective or pathological result might pose a psychological burden for the pregnant woman or the couple.
In particular, if the first trimester screening is followed by secondary tests, such as complicated and lengthy genetic analysis the mental strain might be considerable.
In these situations, we will organize contact to institutions, which provide psychological support.

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