Pränatalzentrum
A.D. WIEN

Between 12 (11+3) and 14 (13+6) weeks pregnant

Information about Screening 12

Nuchal translucency scan, combined test and NIPT

Almost all children (98%) are born healthy.
It is rare for abnormalities to occur.

When and how?

This test can be performed **between 12 (11+3) and 14 (13+6) weeks pregnant** (or if the crown-rump length is between 45 mm and 84 mm). **The optimum time is from 13 weeks pregnant (12+0).** The scan is usually performed through the abdominal wall but if visual conditions are unfavourable (e.g. strong abdominal wall, awkward position of baby, etc.) then through the vagina.

The probability of having a baby with certain chromosomal disorders increases with increasing age of the pregnant woman.

Common chromosomal disorders are
- **Trisomy 21** (Down's syndrome),
- **Trisomy 18** (Edwards' syndrome) or
- **Trisomy 13** (Patau's syndrome).

What is measured?

Many unborn babies with a chromosomal disorder reveal **abnormalities in the ultrasound scan**, including malformations and/or markers such as increased nuchal transparency. **To assess the risk more accurately, all additional markers are examined** (nuchal transparency, nasal bone, tricuspid valve, Ductus venosus).

Evidence of such markers does not necessarily mean that there is a chromosomal disorder but it increases the risk of it, while the absence of such markers lowers the risk.

What does the result mean?

At every S**creening 12 test (nuchal transparency, combined test and NIPT)**, your baby's development is also assessed and, if possible at this early stage of pregnancy, its organs. **About 60% of all serious malformations can be detected by this early screening for malformations.**

The first trimester screening test can never completely exclude the presence of a malformation, disease or chromosomal abnormality in the unborn baby but it reduces the probability if the result is normal. It is a screening test and not a diagnostic procedure.

Chromosomal disorders and genetic abnormalities (e.g. trisomy, microdeletions, etc.) can only be definitely confirmed or 100% excluded through invasive test methods such as **chorionic villus sampling (placenta sample)** from 12 weeks pregnant or **amniocentesis (amniotic fluid test)** from 16 weeks pregnant. However, there is an associated risk of miscarriage of approximately 0.12% to 0.5%.

Nuchal transparency

→ Detection rate for trisomy 21 approximately 80%

The risk of trisomy 21, 18 or 13 being present is calculated through the **combination of the mother's age and all additional markers** (nuchal transparency, nasal bone, tricuspid valve, Ductus venosus).

The result will be discussed in detail with you shortly after the test.

NIPT (non-invasive prenatal test)

→ Detection rate for trisomy 21 > 99%

Based on the **analysis of cell-free DNA in the maternal blood, a particularly reliable and risk-free risk assessment** is carried out mainly for trisomy 21 but also for trisomy 18 and 13.

Possible from 11 weeks pregnant (10+0). In this case, first trimester screening is included at 12 to 14 weeks pregnant. The result is available after approximately 7 to 10 working days. Screening for pre-eclampsia can also be carried out.

Combined test including screening for pre-eclampsia

→ Detection rate for trisomy 21 approximately 90%

Together with maternal age and all additional markers (nuchal transparency, nasal bone, tricuspid valve, Ductus venosus), **two hormones (beta-hCG and PAPP-A) are also measured from a sample of the pregnant woman's blood** and included in the risk assessment.

The result will be discussed in detail with you shortly after the test.

Pre-eclampsia affects approximately 2 to 4% of all pregnancies. It is a complication in pregnancy characterised by high blood pressure and other problems (e.g. protein found in the urine, elevated liver enzymes, the unborn baby's growth is restricted, etc.). Through the combination of different measurements (uterine blood flow, hormone levels from maternal blood, medical history and blood pressure measurement), the personal risk of potentially later developing pre-eclampsia can be assessed.

If there is increased risk, this can be greatly reduced by taking Thrombo ASS 150 mg daily.

I have read, understood and taken note of the different test options, their limitations and potential consequences.

I would like the following tests (please tick those that apply):

- ○ **Nuchal transparency**
- ○ **Combined test** incl. screening for pre-eclampsia
- ○ **NIPT**
- ○ **NIPT** incl. screening for pre-eclampsia

Name of patient

Vienna, (date)

Signature of patient

Signature of doctor

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