Ordination für Pränataldiagnostik
www.ultraschall-csapo.at
Schubertstrasse 16a, 8010 Graz
office@ultraschall-csapo.at
Tel: +43-676-3277-889
Fax: +43-316-231123-5988

Dr. Bence Csapó
Ordination für Pränataldiagnostik
Geburtshilflichen und
Gynäkologischen Ultraschall
www.ultraschall-csapo.at

Non Invasive Prenatal Test (cell free fetal DNA test) for trisomy 21, trisomy 18 and trisomy 13

The risk of your baby having any form of chromosomal anomaly is small. The majority of developmental anomalies are not of genetic origin (about 90%). The statistical risk for trisomies depends on your age. Combined Test is one of the best methods to assess the risk of fetal developmental and chromosomal anomalies in the first trimester of pregnancy. It detects about 93% of embryos with trisomy 21 (formerly also known as Down's syndrome), and most fetuses with trisomy 18 and trisomy 13. For many families trisomy 21 is of special interest, as it is not only the most common chromosomal anomaly, but also because people with this condition have a close to normal life expectancy. The **Combined test unfortunately cannot identify all fetuses affected by trisomy 21 (detection rate 93%, for a false positive rate of 2,5%)**

NIPT is available since 2012 and can detect sings of trisomies from a small maternal blood sample better that a Combined Test. This test is also called Cell-free-fetal DNA or NIPT (non-Invasive-Prenatal-Test). Several companies offer this screening. Common to all is the very high negative predictive value (**the tests can exclude trisomy 21 with a certainty of over 99%**). It can correctly identify more than 99% of trisomy 21 fetuses, over 96% of trisomy 18 fetuses and over 92% of trisomy 13 fetuses.

Limitations of NIPT:

- **NIPT cannot be used instead of the Combined Test!**
- **A detailed ultrasound evaluation of the pregnancy can provide important information NIPT cannot assess!**
- **Over 90% of fetal developmental anomalies cannot be detected by NIPT**
- If not combined with a detailed sonographic evaluation of the fetal anatomy (as done in a combined test), **up 24% of clinically relevant severe fetal anomalies will be missed**!
- **A detailed ultrasound assessment (not compulsory part of NIPT) can identify about 50% of all developmental anomalies identified later in pregnancy already at 11-14 weeks.**
- **Detection of major fetal structural anomalies (only available through ultrasound) makes personal counselling possible.**
- **NIPT does not offer a 100% detection rate** (see above),
- NIPT also has a **false positive rate** ranging **from 1%** for trisomy 21 **to up to 20%(!)** for trisomy 13
- **Screening based on NIPT alone has resulted in an increase of invasive testing rates (amniocentesis = AC) and miscarriages connected with AC in countries where this was introduced.**
- The test is mostly possible (ca. 99,5%) if the mother weighs less than 90kg.
- The **test can fail** and does not give any results in about 3% of all cases.
- At test failure or higher maternal weight a second blood sample may be necessary (at no additional costs for you).
- Even if the companies offer to test **anomalies of the sex chromosomes** and also of certain **microdeletions**, there is **currently no robust information** on how good these results are. High rates of **false positive** results are to be expected, and therefore great caution is recommended! **The governing societies do recommend this type of testing!**
- **In case of abnormal NIPT results (even in the suspected false positives), it is always recommended to verify the results with an invasive test (amniocentesis).**

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When and how is the test done:

- The screening test NIPT is available from week 10+1 (verified by fetal CRL measurement).
- NIPT cannot cause a miscarriage.
- You will receive a detailed counselling (written and verbal).
- A short ultrasound scan will be performed (usually through the abdomen) to verify the fetal size and the heartbeat.
- **It is recommended that a detailed ultrasound assessment of the fetal anatomy be performed (same as the Combined Test).** This is at an additional charge (please see reverse side).
- A small maternal blood sample will be drawn and sent for assessment.
- The screening test is also available for twins, but not for triplets.
- Determination of fetal gender is available for singleton pregnancies, but not for twins.

The NIPT test results are available in 8-10 days. You will be contacted, and a copy of the report will be sent to both you and also to your gynaecologist.

Who benefits most from an NIPT*:*

- Women with an **increased risk for trisomy 21** (or 18, 13, but no other suspected genetic anomalies) based on maternal age or especially after Combined test (recommended cut-off is 1:100).
- Every woman must decide for herself at which risk level she feels uncertain. Some women are happy to not to have further testing at a risk 1:20, while others want more information even at better than 1:1000. It is a personal decision.
- Until NIPT you had to have a needle test (invasive test = amniocentesis) if you wanted more information. An amniocentesis has a small **miscarriage risk of 1:1000**
- NIPT provides you with further information with no risk of miscarriage.
- **NIPT is NOT a diagnostic test – it does not guarantee a healthy baby!**
- The drawbacks of NIPT are the price and that it only works well for trisomy 21, 18 and 13.

The screening test NIPT costs €600.
A short ultrasound is included to measure the size of the embryo and to verify the heartbeat.
A detailed ultrasound assessment of the fetal anatomy is recommended (at additional cost).
The price of the examination is due at the time of the visit (cash or electronic payment is available)

Please consider:
Recent research has proven, that the risk of severe preeclampsia (before week 34) can be evaluated at 11-14 weeks (95% detection rate). This occurs in 3-8% of all pregnancies (most common in the first pregnancy (5-10x risk!)) and is the most common cause of severe maternal and fetal complications during pregnancy.
Preeclampsia is far more common than fetal chromosomal anomalies.
A screening is available at 11-14 weeks, but not later!
A treatment is available to reduce the risk and protect both mother and fetus from severe complications.
The treatment is only effective if started before week 16!
It is strongly recommended to combine a screening test for fetal anomalies with a screening for preeclampsia, especially in first pregnancies!

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Consent for NIPT

I have received for me adequate information about NIPT and I received adequate answers to all my questions. I request the following test:

Singleton pregnancy:

☐ *NIPT (cf-fDNA Test)* ***without*** *detailed ultrasound:* € 600 – (fetal gender optional)

☐ *NIPT (cf-fDNA Test)* ***with*** *detailed ultrasound:* € 750 – (fetal gender optional)

Twins:

☐ *NIPT (cf-fDNA Test)* ***without*** *detailed ultrasound:* € 600 – (fetal gender not available)

☐ *NIPT (cf-fDNA Test)* ***with*** *detailed ultrasound:* € 800 – (fetal gender not available)

Date: _____ Signature: _____

p.s.
If the test cannot be evaluated due to reasons outside our control (e.g. postal strike, or laboratory error) a repeat test can be offered at no additional charge for you. If you decide against a retake, an administration fee of €90 will be retained and the difference will be reimbursed to you.