## Is the child‘s gender automatically analysed?

No, this is only optional included in the test and offered as a self-paid service. If you choose this option, the result will be sent to you in accordance with the provisions of the Genetic Diagnostics Act after the 14th week of pregnancy.

## When is NIPT covered by health insurance?

As a screening test that is not generally recommended during pregnancy, NIPT is only paid for by statutory and private health insurance companies if there is an indication from other tests or if your personal situation makes a risk assessment using NIPT advisable after consultation with your doctor.

## What is family life like with a child with a trisomy?

It is not possible to give a generalised answer, as it depends on various factors. On the one hand, the type and severity of the trisomy play a role, and on the other, the support of the family and the interaction of everyone. Early intervention centres, counselling centres and parents‘ associations can challenge or exchange experiences.

## Further information

This information flyer cannot answer all your questions about NIPT. It is only intended to support the advice given by your doctor or a counselling centre. The Genetic Diagnosis Act and the Pregnancy Conflict Act describe your rights to information and counselling.

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## Note

The content of this flyer is for information purposes only. It does not replace an individual medical consultation or examination.

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stamp of doctor‘s office



**Information for pregnant women**

# NIPT
# The Non-Invasive Prenatal Test for trisomy 13, 18 and 21

With sex determination of the foetus on request

LABOR HEIDRICH & KOLLEGEN

## Dear reader,

this flyer provides you with the most important information on non-invasive prenatal testing (NIPT) for trisomies 13, 18 and 21. NIPT – covered as a health insurance benefit since 1 July 2022. You will obtain very reliable information of the risk for your unborn child carrying the chromosomal disorders trisomy 13, 18, and 21, respectively. These genetic changes affect the child‘s physical and mental development to varying degrees. Trisomy 21, so called Down‘s syndrome, is the most common of them.

The most important risk factor for such a chromosomal disorder is the age of the pregnant woman. For example, about 1 in 1,100 pregnancies is affected at age 20, 1 in 300 at a mother‘s age of 35 and 1 in 68 pregnancies in 40-year-old pregnant women.

Together with your doctor, you may discuss and decide whether a blood based NIPT is the rigth option.
Please be aware, that a consultation with your doctor or with us in the laboratory is required in accordance with the German Genetic Diagnostics Act before you decide for the test.

You are also entitled to psychosocial-counselling from pregnancy advice centres. You will also receive support and advise in case of a suspicious result. This does not necessarily lead to a premature termination of the pregnancy. It may also be the basis to prepare for a child with a trisomy.

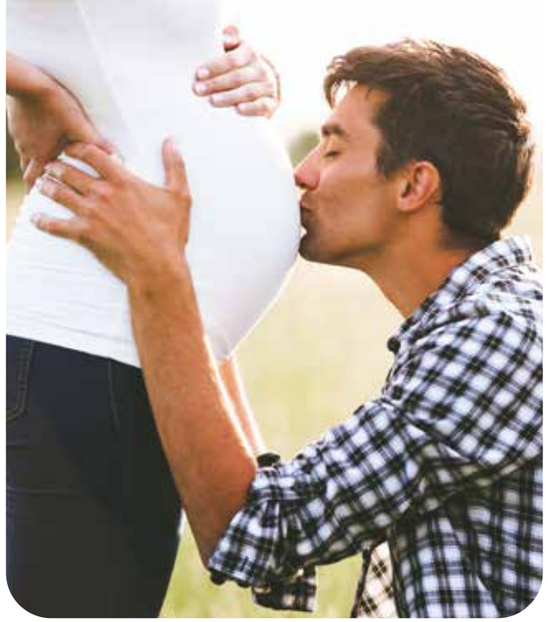
## What are trisomies?

In the case of trisomies, certain chromosomes containing the genetic information are located in the cells of the child threefold instead of twofold and influence the child‘s development in the womb. The risk of trisomy affected babies increases with the age of the pregnant woman.

**Trisomy 13**, also known as Patau syndrome, occurs with an incidence of 1-10 in 10,000 pregnancies. Severe malformations of the heart and brain as well as cleft lip and palate and mental disability will be the result. Most children die in the womb, less than 10 per cent of babies survive the first year.

In the case of **trisomy 18**, Edwards‘ syndrome, the incidence is 2-41 in 10,000 pregnancies. Children with this diagnosis can already have visible malformations of the head, body and internal organs on ultrasound, as well as a mental disability. These malformations usually lead to death in the womb or immediately after birth. Only around 10 per cent with mild disabilities become five years old or older.

**Trisomy 21**, also known as Down‘s syndrome, is the most common type with an incidence of up to 163 per 10,000 pregnancies. Physical symptoms may include congenital heart disease, developmental disorders of the gastrointestinal tract, endocrine disorders, diseases of the skeletal system and visual and hearing disorders. Motor and language development may be delayed. Trisomy 21 in particular has a broad spectrum of manifestations. Some people can be severely physically and mentally restricted, while others will live an almost normal, self-determined life. Life expectancy is about 60 years.



## How does the NIPT works

Starting at the 10th week of pregnancy, the maternal blood contains enough genetic material of the unborn child, so that it can be analysed in the laboratory. Only a blood sample is required for the NIPT, which does not increase the risk of a miscarriage. The analysis of specific DNA sequences of individual chromosomes detects highly reliable additional DNA copies and thus a trisomy can be concluded. This NIPT will not determine whether the unborn child has any other diseases.

## How certain is the NIPT?

In our laboratory we use the Vanadis® method from the manufacturer Revvity. It is considered one of the best (study: Conotte. Comparing 2 non-invasive prenatal testing methods. Am J Ostet Gynecol, 2022). All cases of trisomy 13 and 21 were correctly detected. In the case of trisomy 18, 14 out of 15 cases were correctly detected. (See table below.)

The test is a screening test with risk assessment. The result provides a risk estimate of having a child with a trisomy. Further, so-called invasive diagnostics are therefore carried out to establish a diagnosis if the NIPT result is suspicious. This can be a chorionic villus sampling or an amniocentesis, which can confirm or rule out the risk with more certainty.

In rare cases, no test result can be obtained. The cause may be too low content of fetal DNA present in the sample. Other influencing variables can interfere with the test and lead to incorrect results. However, this is rare. This includes, for example, treatment with heparin (blood clotting inhibitor), a transplant or stem cell therapy in the past, multiple births, a vanishing twin (one twin dies in the womb) or even being severely overweight.

| Chromosomal disorder | Trisomy 13 | Trisomy 18 | Trisomy 21 |
|---|---|---|---|
| Recognition rate | > 99 % | > 93 % | > 99 % |
| False positive rate | 0.1 % | 0.2 % | 0.04 % |

## What are the limits of NIPT for trisomy 13, 18 and 21?

Screening is carried out exclusively for the above-mentioned changes, which represent only a portion of the chromosomal abnormalities or other complications. Regular check-ups or a high-resolution ultrasound scan of the unborn child cannot be replaced by this.

## How and when will you receive the results?

The results will be sent to your doctor within one week. Only they are authorised to inform you of the result. Here it is also possible to obtain only partial results - e.g. only for trisomies 13 and 18, but not for Down‘s syndrome. Legally, the foetus‘ sex may only be communicated after 14+0 weeks‘ gestation.

## Prerequisite for performing the NIPT

According to the German Genetic Diagnostics Act, appropriate counselling and information must be provided by a qualified doctor before genetic tests are carried out. The pregnant woman must consent to the test in writing.