What is the procedure for the PreNata® NIPT test in practice?

1. You receive comprehensive counselling and clarification from your physician according to the Gene Diagnostic Act (GenDG).

2. Your physician takes a blood sample from you.

3. The analysis of your blood sample takes place in our laboratory.

4. The results are transferred to your physician within a few days.

5. Your physician explains the results to you.

Can PreNata® NIPT be performed if you are pregnant with twins?

The test can be performed for a twin pregnancy without any problems. In determining the gender, it can be established if at least one child is male. It is not possible to test for changes to the sex chromosomes (options 3 and option 4). For multiple pregnancies (3 and more foetuses), the test cannot be performed.

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Can PreNata® NIPT be performed after fertility treatment?

PreNata® NIPT can also be performed after fertility treatment without any problems and without limitations.

How reliable is PreNata® NIPT?

The test detects the most common autosomal chromosomal changes (trisomy 13, 18 and 21) with a probability of over 99% and can also exclude them with the same probability. PreNata® NIPT also reliably detects sex chromosome changes and RAAs with 96% probability and CNVs with 74% sensitivity

What do the results mean?

A normal result means that a chromosome change can be almost excluded. If there is an abnormal result, there is a high probability of a chromosomal change. As this is a screening test and the result is not a diagnosis, an abnormal result must be confirmed by further diagnostic procedures. Your doctor will explain the options to you; this usually involves an amniocentesis or an examination of placental tissue (chorionic villus sampling).

Limitations of the PreNata® NIPT

The changes examined represent only some of the possible prenatal chromosomal changes. It is not possible to identify other abnormalities and malformations, or to make statements about the child's state of health. This does not replace the need for regular screening examinations or the obstetric ultra sound examination. Furthermore, mosaics cannot be detected with certainty. The presence of a so-called “vanishing twin” may result in a false positive result. This can also lead to a discrepancy in gender determination.

Test failure

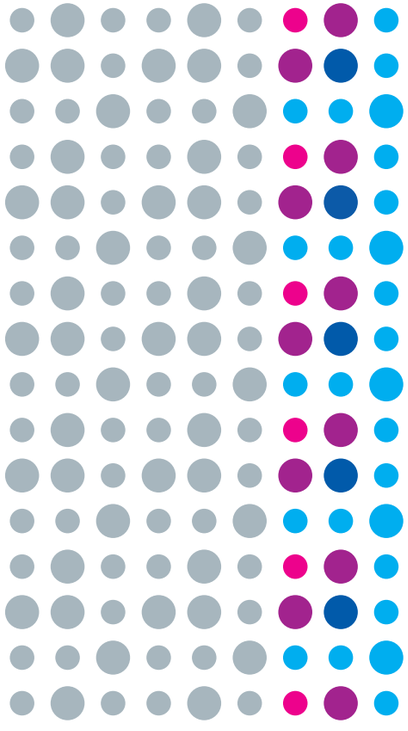
Very rarely, PreNata® NIPT fails to produce a reliable result. If this does occur, a common cause is that the proportion of foetal DNA in the maternal blood is too low. It is then possible to repeat the test free of charge with a new blood sample.

Important notes

In very rare cases (<1%), false negative results can occur. This means that the test gives a normal result, although the child is affected by a chromosome change. Similarly, an abnormal result does not always mean that a chromosomal change is actually present. An abnormal PreNata® NIPT result does not need to lead to a termination of pregnancy, but can be a reason for preparing for a child with a trisomy, for example, by getting confirmation using further tests.

PreNata®

PreNata® NIPT non-invasive prenatal test



You can find further information on PreNata® NIPT at prenata.de

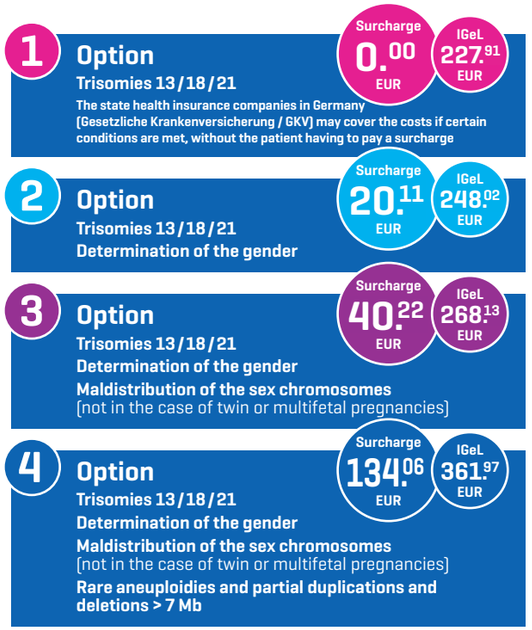
PreNata® NIPT at a glance

The PreNata® NIPT (non-invasive prenatal test) is performed without any risk to the mother or child, to ascertain whether a child is affected by genetic changes that could impair its development. All that is needed is a blood sample from the mother. If desired, PreNata® NIPT can also be used to determine the gender of the child. The examination can be performed from the 10th week of pregnancy.

How does PreNata® NIPT work?

The human genome – our DNA – is contained in the cells in chromosomes. There are usually 23 chromosome pairs per cell. Chromosomes 1–22 (autosomes) and the sex chromosomes (gonosomes) are present twice – one copy each from the father and mother. Women usually have two X chromosomes (XX) and men, an X and a Y chromosome (XY). Chromosome changes, i.e. missing or additional chromosomes (or chromosome parts), can affect development. With the PreNata® NIPT, the child's DNA is examined for chromosome changes using a blood sample from the mother, without risk for mother or child, from the 10th week of pregnancy. Information about the child's DNA is sequenced in the laboratory and it is established whether the child is affected by chromosomal changes. In the case of trisomies, the respective chromosome is present three times instead of two times. In the case of monosomies, however, the affected chromosome is only present once. The children affected usually have clinical abnormalities or health limitations of varying severity. Fundamentally, these kinds of chromosomal changes are very rare, but can also occur in young mothers.

What does PreNata® NIPT cost?



Since 01.07.2022, the non-invasive prenatal test for trisomy 13, 18 and 21 may be paid for as a insurance-covered procedure via your state health insurance (option 1). Speak to your physician to see whether you fulfil the requirements to have the costs covered by your health insurance. Furthermore, in consultation with your doctor, you can additionally opt to have your child's gender determined (option 2) or to test for more extensive chromosomal changes including the determination of the child's gender (options 3 and 4). You have to pay for the cost of procedures which are additional to the insurance-covered procedures yourself (IGeL).

Which chromosomal changes are examined?

The PreNata® NIPT allows you to select from four different options, each with a different scope of examination. The most common trisomies (13, 18, 21) are examined in option 1. Additional options are gender determination, examination of the sex chromosomes, but also the examination of all 22 chromosome pairs (autosomes). However, only those chromosomes are examined that are requested by the choice of examination option. Both the samples and all data collected remain in Germany.

Trisomy 13, 18 and 21

These three trisomies are the most common chromosomal changes in live births.

→Trisomy 13 (Patau's syndrome, frequency ca. 1:5000)
→Trisomy 18 (Edwards' syndrome, frequency ca. 1:3000)
→Trisomy 21 (Down's syndrome, frequency ca. 1:700)

Sex chromosomes

DNA sections of the sex chromosomes X and Y (gonosomes) can also be tested. This can provide indications of the presence of various syndromes in which the sex chromosome are present in a changed quantity, i.e. they are missing or there are more of them (monosomy X; trisomy X, XYY, XXY). These changes are frequently linked to less limiting abnormalities, compared to changes of the chromosomes 1–22 (autosomes). To test the sex chromosomes, select PreNata® NIPT option 3 or 4.

Gender determination

If desired, PreNata® NIPT can also determine the child's gender (options 2, 3 and 4). The notification in accordance with the provisions of the Gene Diagnostics Act is made by your attending physician after the 12th week of pregnancy (14+0 p.m.) if requested.

Genome-wide test of all chromosomes (1–22)

Through the genome-wide examination of all chromosomes, rare maldistributions of complete autosomal chromosomes (rare autosomal aneuploidies – RAAs) can also be identified. These are often so-called mosaics, which means that there are at least two different cell types with different sets of chromosomes. This can affect cells of the child (foetal mosaic) and/or only parts of the placenta (placental mosaic).

When parts of chromosomes are missing (deletions) or present several times (duplications) and thus the number of copies of the affected sections is altered, we refer to copy number variations or CNVs. CNVs occur with a frequency of less than 0.02% in births. They can lead to malformations of the organs and/or developmental disorders. They can be detected with PreNata® NIPT (option 4) from a size of 7Mb.