Eng. - Version 12/12/2025

NIPT CARE

Community for Awareness, Resources, and Education on the NIPT

Every pregnancy is unique and very precious, deserving high-quality prenatal care. As part of this care, you will have the option to take a **non-invasive prenatal test** from the 12th week of your pregnancy onwards. This test is commonly known as the **NIPT**. Nowadays, this test is also referred to as the **non-invasive cell-free DNA screening**. This name gives a better idea of how the test works, and we'll explain it in more detail on this website.

The **NIPT** is a **genetic screening test** that primarily assesses the risk of **trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome),** and **trisomy 13 (Patau syndrome) in your baby**.

This website and the animation movie have been created by the **8 recognized Belgian Centers for Human Genetics**, with the goal of providing clear and accurate information to future parents. The information on this website relates to the NIPT as it is offered in the 8 Belgian genetic centers. When your NIPT is performed in another laboratory, some information might be different. You can discuss with your health care professional where your NIPT will be analysed.

Choosing a NIPT is entirely yours. This test may bring peace of mind, or, in some cases, raise new questions. Take time to watch the animation movie and to read the leaflet and website. If you still have questions, your healthcare professional will be happy to discuss them with you.

1. WHAT IS THE NIPT?

Each cell of our body contains DNA, the genetic material that carries the instructions for how our bodies grow and function. This DNA is organized into structures called chromosomes. Humans have **46 chromosomes**, arranged in 23 pairs. From each pair, we inherit one chromosome from each parent.

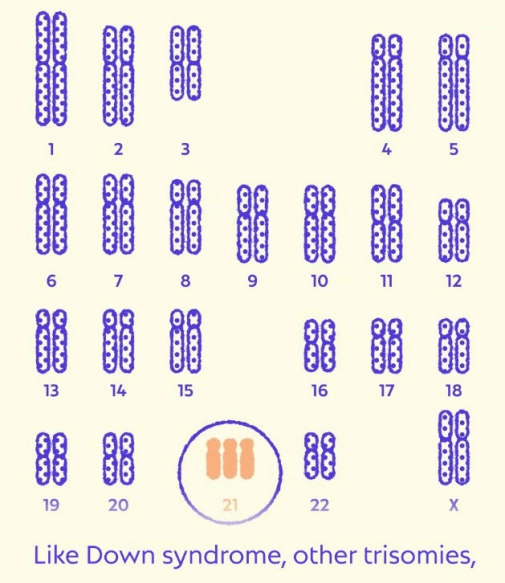
The first 22 pairs, called the **autosomes**, are the same in both men and women. The 23rd pair are the **sex chromosomes**, which determine a person's sex: women have two X chromosomes (XX), while men have one X and one Y chromosome (XY).

Sometimes, there can be a difference in the number or structure of chromosomes. These are called **chromosomal abnormalities**.

- One common type is when a chromosome is missing or there's an extra chromosome. This is known as an **aneuploidy**.

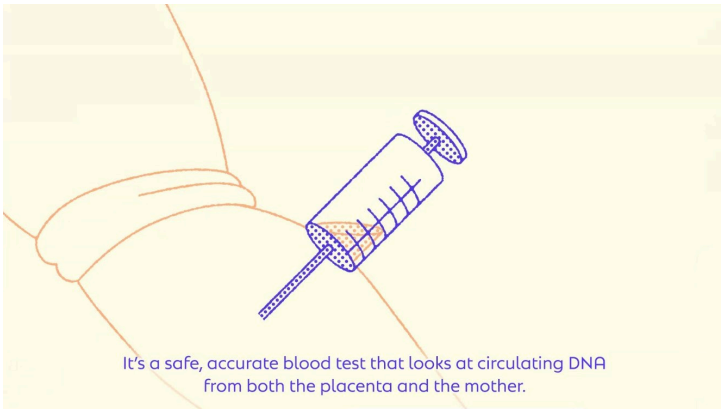
    - In case of an extra chromosome (so three copies instead of the usual two), it is called **trisomy**. A well-known example is trisomy 21, which causes Down syndrome (see figure).
    - A missing chromosome is called **monosomy**. For instance, monosomy X, where a female has only one X chromosome instead of two, causes Turner syndrome.
- In some cases, only a part of a chromosome is missing (deletion or loss) or extra (duplication or gain); this is called a **segmental abnormality**.

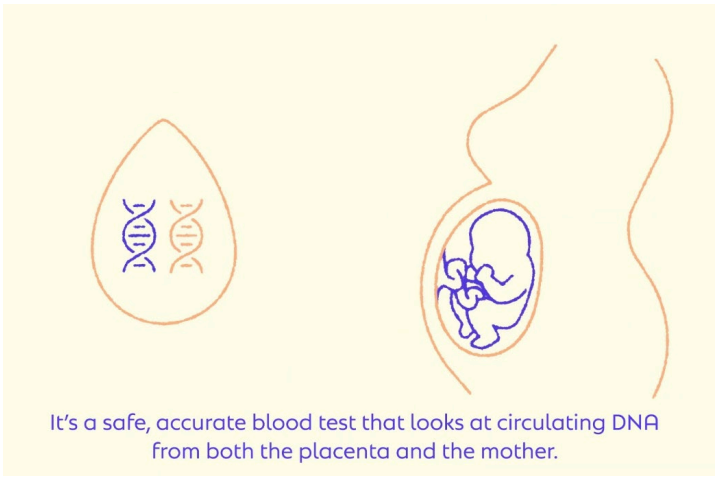
Some of these changes can affect a baby's development, of which some of them can be detected during pregnancy through screening or diagnostic testing.



Example of a karyotype with an extra chromosome 21, or trisomy 21, the genetic cause of Down syndrome

For the NIPT, a blood sample is taken from the pregnant woman. This sample contains, among others, small fragments of DNA that come from different parts of her body, but also DNA fragments of the placenta. These fragments of genetic material are called **'cell-free DNA' or cfDNA**, since they are no longer part of a cell, but float freely in the blood (see figure). The part of the cfDNA that originates from the placenta is examined during NIPT. Since the placenta usually has the same genetic makeup as the baby, this is the type of genetic material we want to look at to check the baby's chromosomes. The amount of cfDNA from the placenta compared to the amount of total cfDNA in the blood stream is called the **fetal fraction**.





For the NIPT, only a blood sample from the pregnant woman is needed. This blood contains small fragments of DNA that come from both the mother's organs and the placenta. These fragments, known as cell-free DNA or cfDNA, are analysed during an NIPT to screen for certain chromosomal abnormalities in the baby.

Using advanced technology called massively parallel sequencing (also known as next-generation sequencing), the amount of cfDNA fragments can be counted (from both mother and baby). If there are more fragments of a certain chromosome than expected, this might mean that there is a trisomy of that chromosome (in the placental cfDNA), too little fragments can indicate a monosomy.

2. WHEN TO PERFORM THE NIPT

The NIPT is a **safe and simple blood test** that you can take at the end of the first trimester of your pregnancy. As early as a few weeks into your pregnancy, cfDNA from the placenta is present in your blood. By the 12th week of your pregnancy, the amount of cfDNA from the placenta is usually high enough to obtain a reliable result.

The first-trimester ultrasound is mandatory before undergoing NIPT. This ultrasound checks for structural abnormalities and measures the nuchal translucency (NT) of your baby. If

any abnormality is detected, NIPT may not be appropriate. In such cases, further prenatal follow-up, genetic counselling and possibly invasive diagnostic tests (like chorionic villus sampling or amniocentesis) are recommended instead of NIPT.

3. WHAT DOES THIS TEST EXAMINE

The NIPT is a genome-wide test. This means that all chromosomes can be examined, but with a primary focus on chromosomes 13, 18, and 21, since trisomies of these chromosomes are the most common and can lead to a liveborn baby. Detecting these conditions early through a NIPT can help you and your healthcare professional to prepare for the next steps, whether that involves further testing, medical planning, or supportive care.

In rare cases, other abnormalities may also be found, such as a trisomy of another autosome, called a rare autosomal trisomy or RAT, or a smaller abnormality of only a part of a chromosome. Since both cfDNA from you and your baby are analysed, it is also possible that an abnormality in your DNA is identified. If this finding is important for your health and/or the health of your baby, this result will be communicated to your healthcare professional so that appropriate clinical follow-up can be offered to you.

3.1. Trisomy 21

A baby with **Down syndrome** has three copies of chromosome 21 instead of two. The main characteristics of trisomy 21 are typical facial features and a mild to severe intellectual disability. There is often a congenital heart defect, but other organ abnormalities can also occur. In some cases, Down syndrome can already be suspected through ultrasound examination (e.g. an increased nuchal translucency). The risk of having a pregnancy with trisomy 21 increases with age of the mother and rises especially after the age of 35.

3.2. Trisomy 18

Trisomy 18 or **Edwards syndrome** is a serious genetic condition caused by an extra copy of chromosome 18. Babies with this condition are usually very small at birth and may have a low birth weight. They often have distinct physical features, such as a small head (microcephaly), a small jaw (micrognathia), low-set ears and clenched fists with overlapping fingers. Their feet may have a rounded shape, known as "rocker-bottom feet." In addition to these physical abnormalities, babies with trisomy 18 often have serious health problems. These can include heart defects, kidney abnormalities, and issues with breathing and feeding. Most children also have severe developmental delay and intellectual disability.

Many of these pregnancies end in a miscarriage or stillbirth. Only about 5–10% of the babies born live beyond their first year.

3.3. Trisomy 13

Trisomy 13 or **Patau syndrome** is a rare and serious genetic condition caused by an extra copy of chromosome 13. Babies with this condition often have multiple physical and developmental differences. They are usually born with a low birth weight and may have a small head (microcephaly), small or missing eyes (microphthalmia or anophthalmia), and a cleft lip and/or palate. Their ears may be shaped differently, and they often have extra fingers or toes (polydactyly). Many babies with trisomy 13 also have severe brain abnormalities, such as holoprosencephaly, where the brain does not divide properly. Heart defects are also very common. Other possible issues include kidney problems, abdominal wall defects (like omphalocele), and poor muscle tone.

Trisomy 13 is life-threatening. Many pregnancies end in a miscarriage, and most babies who are born alive do not survive beyond the first few weeks of life. Only 5% to 10% of babies with a trisomy 13 live past their first year.

3.4. Sex of the baby

Most parents are eager to learn the sex of their baby as early as possible, and NIPT offers this option. However, it is important to realize that this is not the main purpose of the test. NIPT is primarily designed to screen for chromosomal conditions, not to determine the baby's sex.

While the sex of your baby can be predicted with >99% certainty, please remember that this is a screening test. Therefore, it is recommended to confirm the sex later in pregnancy during an ultrasound examination.

At most genetic centers, you can choose whether or not to know the sex of your baby through NIPT. If this is the case, you simply indicate your preference on the consent form before the test is done.

3.5. Rare autosomal trisomy or RAT

In rare cases, NIPT can detect a trisomy of one of the other autosomes, called a rare autosomal trisomy or RAT. Although these rare abnormalities can be present in the baby, mostly they are only present in the placenta, a situation that is called **confined placental mosaicism**. In these cases, the genetic makeup of the placenta and the baby is different. The cause of this difference initiates during the very early stages of pregnancy. Important to know is that certain trisomies in the placenta can cause the placenta to function less efficiently. Therefore, if an increased risk for a RAT is found by NIPT, your healthcare professional can recommend extra monitoring or follow-up tests to ensure your pregnancy is progressing well. Also, an amniocentesis can be performed to exclude the presence of the chromosomal abnormality in your baby. If the baby turns out to be unaffected, your pregnancy will still be monitored more closely to detect any possible complication early, for yours and your baby's wellbeing.

3.6. Smaller chromosomal abnormalities

In very rare cases, NIPT can detect a small change in a part of a chromosome, known as a **segmental abnormality**. These changes are more difficult to detect than an entire extra or missing chromosome, because they involve only a small part of a chromosome. The ability to pick up these smaller differences depends on several factors, one of the most important being the fetal fraction (the fraction of cfDNA from the placenta present in your blood). The higher the fetal fraction, the more accurate these subtle changes can be detected during NIPT. Although very rare, these findings can be important and may lead to further diagnostic testing or closer monitoring during pregnancy.

3.7. Center specific investigations

3.7.1. Sex chromosomal aneuploidies

NIPT performed in some genetic centers can also look at abnormalities of the sex chromosomes (X and Y). These so-called sex chromosomal aneuploidies are caused by an additional or missing sex chromosome. Most of these conditions do not cause serious health problems, which is why they are not routinely reported. However, there are two conditions, **Turner syndrome** (female with only one X chromosome) and **Klinefelter syndrome** (male with one extra X chromosome), that can affect a child's physical and psychological development. An early diagnosis of these two syndromes can help guide treatment and support from a young age onwards.

Because of this advantage, some genetic centers offer the option to screen for these two conditions during NIPT. If this option is available, you can choose whether or not you want to receive this information by indicating your preference on the consent form before taking the test. You can look at the website of each genetic center to know where this additional analysis is offered.

Turner syndrome

Turner syndrome or monosomy X occurs in about 1 in 2,000 newborn girls. Girls or women with Turner syndrome have 45 chromosomes instead of 46, as one X chromosome is missing.

This syndrome has a very variable clinical appearance. In some cases, Turner syndrome is already suspected during pregnancy through an abnormal ultrasound (e.g. fluid collection, heart anomaly). Girls with Turner syndrome usually appear normal at birth, but they have a higher risk of congenital abnormalities (such as heart defects, kidney abnormalities and lymphedema). Signs often occurring during development include recurrent ear infections with a risk of hearing loss, growth delay for which early growth hormone treatment is indicated, absence of spontaneous puberty and fertility problems. Intellectual development is generally normal. However, there is an increased risk of neurodevelopmental difficulties (such as learning difficulties and problems with motor skills) or psychosocial vulnerability. It is important to emphasize that the characteristics vary from person to person and not all features are always present or equally pronounced.

If an increased risk of monosomy X is found by NIPT, multidisciplinary care is offered through the Center for Medical Genetics and the Pediatric Endocrinology department before and after birth, so that you and your child receive the best personalized follow-up.

Klinefelter syndrome

Klinefelter syndrome occurs in about 1 in 500 boys. Boys and men with this syndrome have one extra X chromosome, which is why the condition is also known as 'XXY karyotype'.

Also, this syndrome has a very variable clinical appearance but cannot be detected on ultrasound during pregnancy. Boys with this chromosomal abnormality are usually taller, have an increased risk of a delayed or less pronounced puberty and infertility. At adult age, metabolic problems such as thyroid problems, diabetes, and osteoporosis occur more frequently. Intellectual development is generally normal, but there is a higher risk of neurodevelopmental difficulties (such as delayed language development and/or learning difficulties) and psychosocial vulnerability. There is evidence that early intervention has a positive effect on psychomotor development. This condition, however, has a variable phenotype and is often discovered in adult men by chance.

If an increased risk of an extra X chromosome in a male baby is found by NIPT, multidisciplinary care is offered through the Center for Medical Genetics and the Pediatric Endocrinology department before and after birth, so that you and the child receive the best personalized follow-up.

3.7.2. Specific microdeletion syndromes

The NIPT can also identify certain microdeletion syndromes. This is done using the same data generated using the standard method but using an additional analysis method looking in more detail to specific chromosomal regions. However, the NIPT cannot detect all cases of rare microdeletion syndromes, and mostly, these conditions are extremely rare. Also, an invasive procedure is obligated after such an abnormity is suggested through NIPT.

Children or people affected with one of these syndromes mostly show moderate to severe intellectual disability, developmental problems and specific facial characteristics, next to some syndrome-specific outcomes. The microdeletion syndromes that can currently be detected via this additional test method are:

- 22q11 deletion syndrome
- Prader-Willi/Angelman syndrome
- Smith Magenis syndrome
- 8p23.1 deletion syndrome

If such an abnormality would be detected, your healthcare professional will inform you and offer you the appropriate multidisciplinary follow-up. You can look at the website of the genetic centers to know where this additional analysis is offered.

3.8. Maternal chromosomal abnormalities

The cfDNA in your blood originates for the largest part from cells of your own body, and not your baby's. Consequently, your cfDNA is also analysed. It is possible that you carry a chromosomal abnormality that can be detected by NIPT. If such a finding is important for your health and/or for the health of your baby, this will be communicated to your referring healthcare professional. In rare cases, an amniocentesis may be necessary to determine whether your baby also carries the abnormality.

In extremely rare cases, NIPT can detect cancer in the mother. This is because cancer cells also can release cfDNA into the bloodstream. If these cancer cells display large chromosomal abnormalities, this can be visible in the genetic pattern of the NIPT. However, note that only a very small number of cancer types can be detected by NIPT. If the test shows a strong suspicion of cancer, you will be informed about this, so that rapid follow-up and multidisciplinary care can be offered.

4. ADVANTAGES AND DISADVANTAGES

4.1. Advantages

This test is a non-invasive test that only requires a blood sample. Therefore, there is no risk of a miscarriage. This is in contrast with so-called invasive prenatal tests, such as an amniocentesis and chorionic villus sampling, that carry a small risk of miscarriage (0.1–0.5%).

This test has a sensitivity of nearly 99% for detecting trisomy 21. This means that 99 out of 100 children with Down syndrome can be identified during pregnancy with this test. The test is also very reliable for detecting the other two common autosomal abnormalities, trisomy 13 and trisomy 18.

4.2. Disadvantages

While NIPT is a highly accurate screening tool, it is important to understand its limitations. First and foremost, it is a screening test and not a diagnostic test. This means it can estimate the risk of certain chromosomal conditions, but it cannot provide a diagnosis. If the NIPT shows something unusual, additional diagnostic testing is recommended. This includes an amniocentesis, in which some amniotic fluid is taken to examine your baby's DNA more directly. A detailed expert ultrasound will also be suggested to closely monitor your baby's development.

NIPT is not suitable to detect all genetic conditions. This means that **even if your result is normal, it does not guarantee that your baby has no genetic or developmental disorder**. The following genetic conditions can for instance **not be detected by NIPT**:

- **Most small chromosomal abnormalities** (so-called microdeletions or microduplications): these are tiny missing or extra pieces of DNA. These changes are often too small to be detected or the amount of the affected cfDNA in the blood can be too low to be measured accurately.
- **Monogenic disorders** (conditions caused by small changes or mutations in a single gene, such as cystic fibrosis or fragile X syndrome): these conditions are caused by a change in a single gene, not a whole (or part of a) chromosome. NIPT is designed to look for extra or missing chromosomes, not single changes in individual genes.
- **Chromosomal rearrangements (e.g. reciprocal and Robertsonian translocations):** If one of the parents carries a balanced chromosome rearrangement (called a translocation), the baby may inherit an unbalanced version of it, which can lead to serious developmental problems. The NIPT is not the correct test to analyse this during pregnancy. Instead, an invasive test, such as amniocentesis or chorionic villus sampling (CVS), will be recommended to look directly at the baby's chromosomes and confirm whether such an unbalanced rearrangement is present.

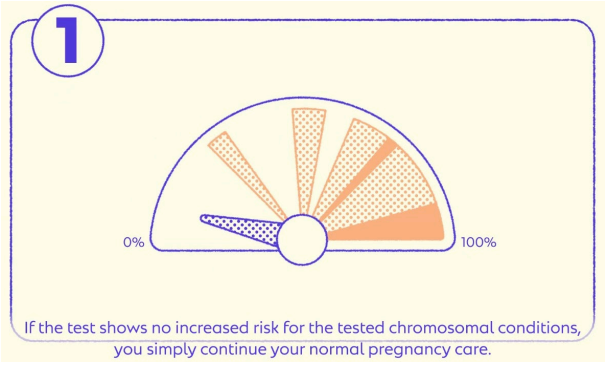
If NIPT is not recommended in your situation, perhaps due to a condition described above, your healthcare professional will explain why. In these cases, an invasive procedure (chorionic villus sampling or amniocentesis) may be the appropriate test.

5. YOUR NIPT RESULT

In most cases, the result is available within 7 to 10 calendar days after the lab receives your blood sample. Ask your healthcare professional for more specific information about how your result will be reported.

These are the possible results:

5.1. Low risk



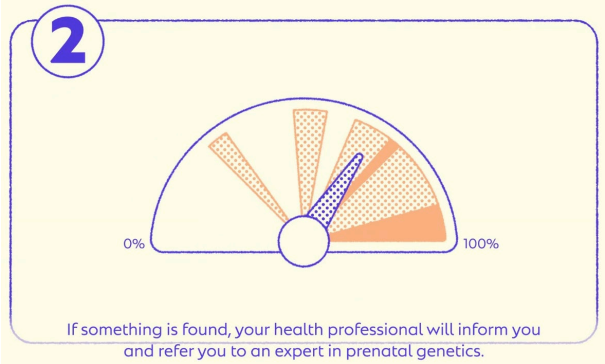
A low risk result means that there is no increased risk for trisomy 13, 18 or 21. When additional testing is performed, it also shows no indication of other detectable chromosomal abnormalities.

The NIPT is highly reliable for detecting trisomy 21, trisomy 18, and trisomy 13. However, it is important to remember that this is a screening test, not a diagnostic test. This means that

while a normal result is very reassuring, it cannot guarantee with 100% certainty that the baby has no trisomy 21, 18 or 13. Please also note that the reliability for detecting the rare autosomal trisomies, sex chromosomal and/or smaller chromosomal aberrations is lower (but the risk that your baby carries one of these aberrations is also very low).

5.2. Increased risk for a chromosomal abnormality

5.2.1. Increased risk for trisomy 13, 18 or 21



If your result shows an increased risk for a trisomy 13, 18 or 21, it is an indication that your baby can have the detected abnormality, but it is **not a definitive diagnosis**. To confirm the result and to provide a clear answer, an invasive diagnostic test via an amniocentesis is required to directly examine your baby's genetic material. This additional test will provide a definitive diagnostic answer about your baby's DNA.

5.2.2. Increased risk for another chromosomal abnormality

NIPT can also detect a chromosomal abnormality beyond the common trisomies (13, 18, and 21). Most often, this concerns a rare autosomal trisomy or RAT. In rare cases (if performed), this can also include a smaller chromosomal change or a sex chromosome abnormality.

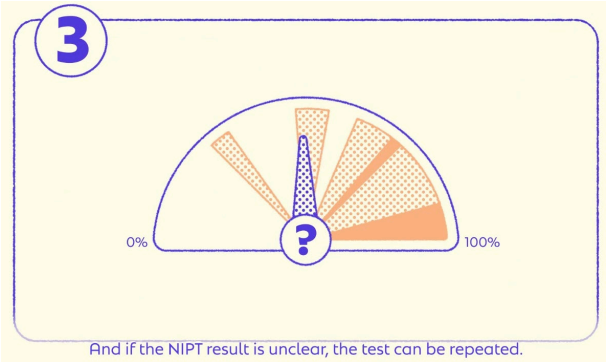
If the detected abnormality is important for the health of your baby, this will be reported to your healthcare professional. Subsequently, you will be informed about the abnormal result and the next steps will be discussed. This most likely will be a referral to an expert in prenatal diagnostics, with an expert ultrasound and /or an amniocentesis for further evaluation.

5.2.3. A chromosomal abnormality in the pregnant person

In rare cases, NIPT may pick up a chromosomal abnormality in your DNA, rather than in your baby's. If such a finding is considered relevant for your own health and/or for the baby's development, your healthcare professional will inform you and explain what it means.

With NIPT, it is not possible to determine whether your baby also carries the same abnormality. However, it is not always necessary to investigate this during your pregnancy. You will be guided through the necessary steps by your healthcare team and an expert in clinical genetics.

5.3. Unclear result



In about 3 to 5% of cases, the result is **inconclusive or uninterpretable**. This means the test could not provide a clear answer. One common reason is that there is not enough placental cfDNA in the blood sample (i.e. when the fetal fraction is too low). One reason for a fetal fraction being too low is an increased BMI. Alternatively, the quality of the data may not be sufficient to give a reliable result.

In rare cases, it is not possible to determine the sex of you baby accurately.

An unclear result does not necessarily mean that there is an increased risk of an abnormal result. In most cases, the presence of a trisomy 21, 18 or 13 can be excluded by repeating NIPT on a second blood sample a little later in pregnancy. This second analysis is free of charge on condition that it is performed in the same genetic center as your first test. You can discuss with your healthcare professional to have the test repeated and/or to discuss whether additional ultrasound follow-up is appropriate.

In most cases, a second test on a new blood sample does provide a reliable result. If the second NIPT is also inconclusive, further follow-up will be discussed. In specific cases (around 20 weeks of pregnancy and with normal ultrasound findings), a third analysis can be performed. In other cases, an invasive test (amniocentesis) may be indicated to receive more conclusive information about the genetic makeup of your baby.

In some cases, repeating the test is not useful. In this case, your health professional will then discuss the other options with you.

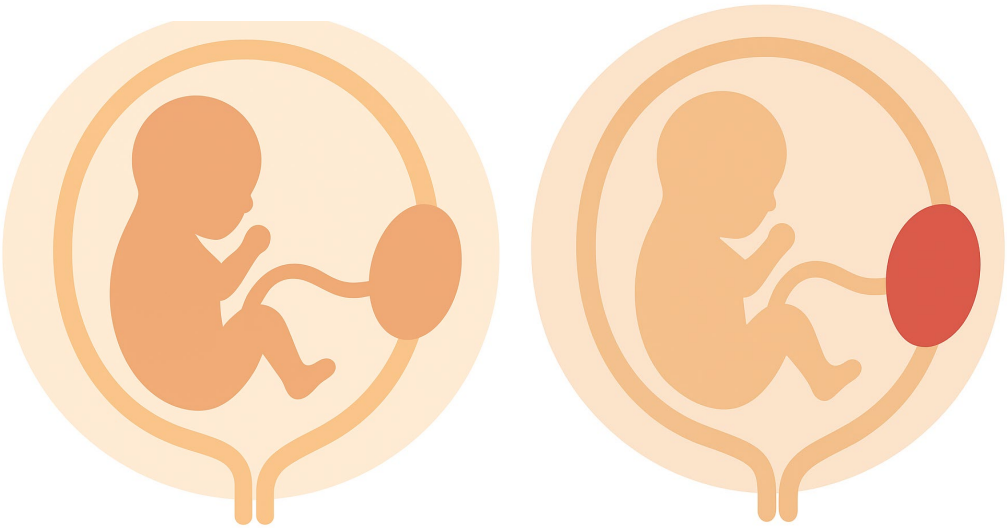
6. FALSE POSITIVE AND FALSE NEGATIVE RESULTS

6.1. False positive result

A result is considered false positive when an abnormal NIPT result is not confirmed in amniotic fluid or in your baby.

Possible explanations are:

- **Confined placental mosaicism** (see figure): As mentioned before, the NIPT analyses cfDNA from the placenta. Most of the time, this DNA matches the baby's DNA. But in rare cases, there can be a difference due to a genetic error that happened very early in pregnancy. If the error is present only in the cells of the placenta and not in your baby's cells, this is called confined placental mosaicism and will lead to a false positive result. For this reason, an amniocentesis is always highly recommended when the NIPT shows an increased risk for a chromosomal abnormality. It is known that certain trisomies in the placenta can cause the placenta to function less efficiently. So, even if the baby doesn't have the genetic condition, changes in the placenta can increase the risk of pregnancy complications, such as **poor growth of the baby and/or preterm birth**. That's why your healthcare professional will monitor your pregnancy more closely if confined placental mosaicism is suspected.



*In the left situation, the **normal NIPT result** correctly reflects the baby's genetic health. Both the placenta and the baby have a normal set of chromosomes.*

*The right image shows **confined placental mosaicism**. A chromosomal abnormality is present in the placenta, but the baby not in the baby. This can lead to a **false positive NIPT result**, since the test analyses cfDNA from the placenta. This situation is extremely rare.*

(made with ChatGPT as example)

- **A chromosomal abnormality in your DNA**: If you carry a chromosomal abnormality that can be detected by NIPT, this could be misinterpreted as being from the baby. However, in most cases, the lab can recognize this during the analysis and will avoid confusion.
- **Vanishing twin pregnancy** (original twin pregnancy in which one baby deceased early in pregnancy): The presence of a vanishing twin with a chromosomal abnormality can cause misinterpretation since the test cannot determine if the surviving baby carries the abnormality as well.

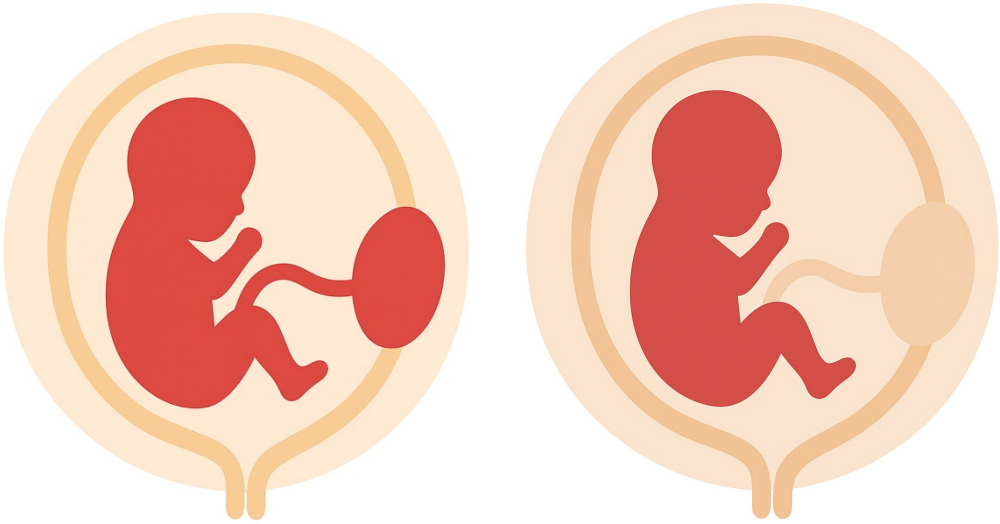
In these cases, an amniocentesis will provide you with the correct (diagnostic) result. Your healthcare professional will subsequently discuss if, and what kind of specific follow-up is still recommended for your pregnancy.

6.2. False negative result

Despite the high reliability of NIPT, there is still a very small chance of a false negative result. This means that the test did not indicate an increased risk for trisomy 13, 18, or 21, but your baby is, at a later stage, diagnosed with the condition.

There are several possible reasons:

- **Confined fetal mosaicism** (see figure): This is similar to confined placental mosaicism, but in this case, the abnormality is present only in your baby and not in the placenta. Since NIPT analyses cfDNA from the placenta, this test may miss an abnormality in your baby if the placenta is normal. This situation is however extremely rare.



*In the left situation, the **abnormal NIPT result** correctly reflects the baby's genetic health. Both the placenta and the baby have the same chromosomal abnormality.*

*In the right situation, **confined fetal mosaicism** occurred. A chromosomal abnormality is present in the baby (or fetus), but not in the placenta. This can lead to a **false negative NIPT result**, since the test analyses cfDNA from the placenta. This situation is extremely rare.*

- **Low fetal fraction**: In this situation, there is too little cfDNA from the placenta present in your blood to obtain a reliable result. A low fetal fraction can occur if the test is performed too early in your pregnancy or in cases of maternal obesity. If, during analysis, there are concerns about a fetal fraction being too low, new blood sampling will be proposed to repeat the test later in pregnancy.

7. TWIN PREGNANCIES

7.1. Monozygotic (identical) twins

A monozygotic twin pair originates from one fertilized egg (oocyte). They can share one placenta (called **monochorionic**), or each have their own placenta (**dichorionic**). Monozygotic twins are almost always genetically identical. Therefore, the result of NIPT is very likely to apply to both babies.

7.2. Dizygotic (non-identical) twins

In a dizygotic twin pair, two oocytes have been fertilized and two placentas are present. Both placentas shed their DNA in the blood. Although it is possible that more cfDNA from one placenta is present than from the other, it has been proven that the chance of missing an abnormality in a dizygotic twin pregnancy is not higher than in an identical twin pregnancy or a singleton pregnancy (with only one baby). This means that NIPT can also be reliably performed in dizygotic twin pregnancies.

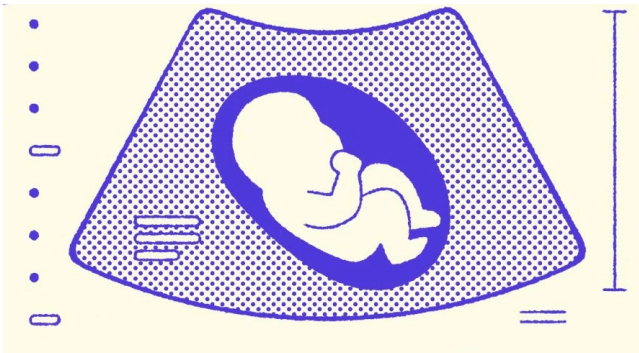
7.3. Vanishing twin

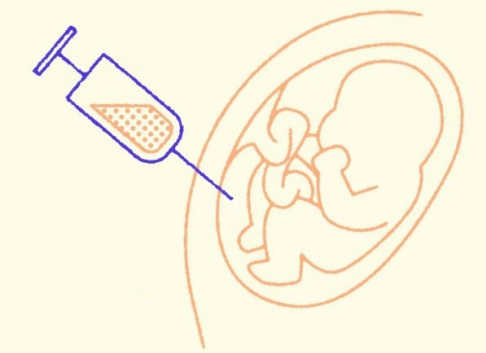
A vanishing twin is a member of an original twin pregnancy that stops developing early in pregnancy. The cfDNA from the vanishing twin can still circulate in the bloodstream for weeks to months during the pregnancy. If NIPT indicates a chromosomal abnormality in this situation, it cannot be determined whether the abnormality originates (only) from the vanishing or (also) from the surviving twin. In this situation, an amniocentesis is recommended to confirm that the surviving baby has no chromosomal abnormality.

8. FOLLOW-UP AFTER AN ABNORMAL RESULT

If the result of your NIPT shows a possible abnormality, it is important to remember that this test is a **screening test, and not a diagnosis**. This means it indicates a higher chance of a chromosomal condition, but it cannot tell with 100% certainty whether your baby actually has the chromosomal abnormality. That is why your healthcare professional may recommend an **expert ultrasound examination** at a specialized center, also known as a **tertiary center**. This type of ultrasound is done by specialists who have advanced training and experience in detecting subtle signs of structural problems in the baby. During the examination, that is completely harmless for you or the baby, the specialist will carefully check your baby's development.

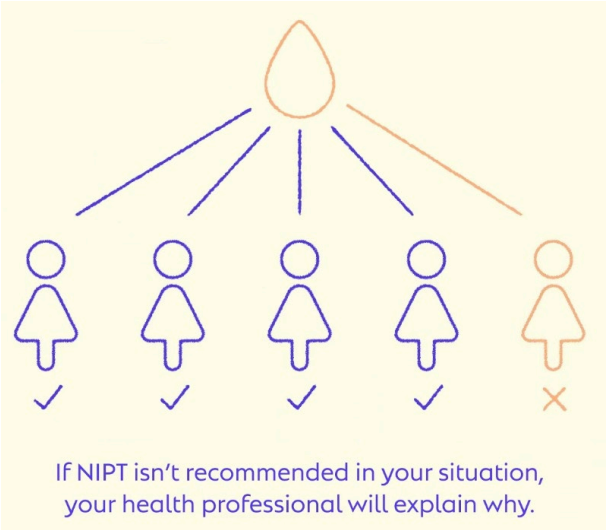
Following the expert ultrasound, an **amniocentesis** might be offered. During an amniocentesis, a small amount of amniotic fluid is taken from your uterus using a thin needle inserted through your belly. An ultrasound is used to guide the needle safely. This amniotic fluid contains cells from your unborn baby which can be tested to check for chromosomal abnormalities. The procedure is performed at a center for prenatal diagnosis. While this invasive test carries a small risk of miscarriage, about 1 to 5 in 1,000 cases (0.1-0.05%), the risk is almost non-existent when being carried out at an expert center. Moreover, it can provide almost complete certainty about whether your baby has the abnormality detected with NIPT.





During a **chorionic villus sampling** (CVS), a small sample of tissue from the placenta is taken. Although CVS can detect many of the same conditions as during an amniocentesis, it is not recommended as a follow-up after an abnormal result from NIPT. This is because both tests analyse DNA from the placenta, not directly from the baby. In case of a confined placental mosaicism, a certain chromosomal abnormality may be present only in the placenta but not in the baby. In such cases, CVS might show the same abnormal result as the NIPT, even if the baby is completely healthy. To avoid this uncertainty, amniocentesis is mandatory as follow-up test after an abnormal NIPT result, as this gives a more accurate picture of the baby's actual genetic makeup.

9. IN WHICH CASES IS THE NIPT NOT RECOMMENDED?



In some cases, NIPT is not recommended. For example, if the 12-week ultrasound shows an abnormality in your baby, which could be suggestive of a genetic condition. Important to know is that several familial genetic conditions cannot be detected by NIPT. In these situations, a diagnostic and more detailed genetic test is recommended on amniotic fluid or chorionic villi.

NIPT cannot be offered if you had a recent organ, blood or tissue transplant or stem cell therapy. These treatments introduce foreign DNA into your blood, which can interfere with the test. The presence of extra DNA from a donor can render the results unclear or misleading.

If NIPT is not an option for you, your healthcare professional will guide you toward the most accurate and appropriate testing options to ensure the best care for you and your baby.

Some medical conditions or therapies can influence the NIPT results. Therefore, it is important to mention to your physician when you fill out the consent form that:

- a specific chromosomal abnormality that runs in your family;
- you have (had) cancer;
- you have an auto-immune disease (such as Crohn's disease);
- you are under heparin therapy.

In this way, this information can be taken into account while analysing your NIPT result.

10. INFORMED CONSENT

A written consent is needed before performing the NIPT.

11. COST

The NIPT is reimbursed if you are a member of a Belgian service for public health insurance. The amount you pay yourself is around 9 euros. This reimbursement is provided for every pregnant woman, once per pregnancy, regardless of her age or family history, but only from the 12th week of pregnancy onwards.

The costs of the consultation with the health professional and the ultrasound are not included in this amount.

If you are not affiliated with a Belgian health insurance service, you will have to pay the cost of the test and a consultation yourself.

12. ACCREDITATION

The NIPT is performed at one of the 8 recognized Belgian Centers for Human Genetics. This is a validated test that has been published in several scientific journals. This test is included in the scope of accreditation according to ISO15189 quality standards through BELAC (215-MED).

In addition, all departments offer multidisciplinary expertise needed to ensure accurate interpretation and follow-up of the NIPT, in accordance with national guidelines from the Belgian Society of Human Genetics (BeSHG) and the recommendations of the Belgian Advisory Committee on Bioethics (Opinion No. 66 and No. 76).

13. USE OF RESIDUAL MATERIAL AND GENETIC DATA

The Belgian Centers for Human Genetics are committed to continuously improve the quality of their care and testing. The residual material and genomic data obtained after NIPT can be used for validation, internal quality control or research purposes (for example, optimization of NIPT and new developments) in line with the general policy at the respective center where you had your test carried out. This information is also mentioned on the consent form.