National Institute for Public Health
and the Environment
Ministry of Health, Welfare and Sport

Testing for chromosomal abnormalities and physical abnormalities

The **NIPT** and the **20-week ultrasound**



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Prenatal screening

You are pregnant. You can check the health and development of the baby you are carrying by getting the NIPT and 20-week ultrasound. This is called prenatal screening.

What tests are available?

There are two types of prenatal screening:
1. A blood test to check for chromosomal abnormalities. This is called noninvasive prenatal testing: the NIPT.
2. A scan to check for physical abnormalities: the 20-week ultrasound.

Which tests can you have and when?



from week 12

NIPT

test for chromosomal abnormalities



around week 20

20-week ultrasound

test for physical abnormalities, about halfway through the pregnancy

Making a choice

When you are pregnant, you go to see a midwife. Your midwife asks if you want to know more about the NIPT and the 20-week ultrasound.

There are two options:

1. You would prefer not to know. That means you will not get any information and you will not have the tests.
2. You do want to know. You will have a long talk with the midwife. After this, you decide if you want to have one or more tests:
   - NIPT: yes or no
   - 20-week ultrasound: yes or no

Here are some questions to consider carefully:

- How much do you want to know about your child before it is born?
- What would it be like for you to live with a child with abnormalities? (It could be a chromosomal abnormality such as Down syndrome, or a physical abnormality?

In this leaflet, we talk about seeing the midwife. Other people may also help you through your pregnancy and childbirth: a gynecologist, obstetrician, nurse, physician assistant, or other care provider.

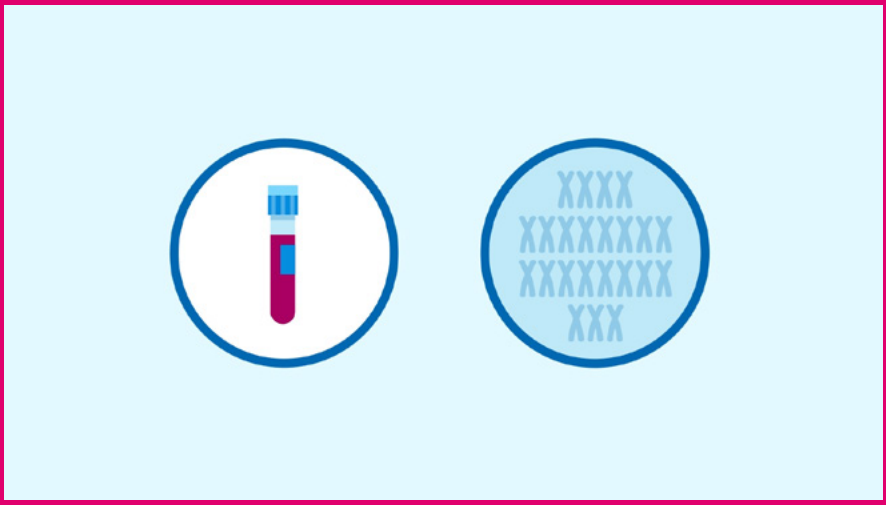


Do you want to have that talk?
Tip: Visit www.screening-cn.com/en/niptseo before you go to the appointment. You can read about the tests there. There are also short videos about the tests.

The NIPT

Noninvasive prenatal testing (NIPT) is a blood test to check for chromosomal abnormalities in an unborn baby. The NIPT involves giving a blood sample. Your blood is tested for chromosomal abnormalities that could have serious consequences for the child's health, such as Down syndrome.

Watch the video about the NIPT



How does the NIPT work?

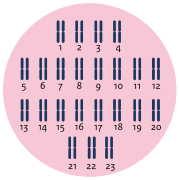
The NIPT involves giving a blood sample. You can have blood drawn from week 12 of your pregnancy. Your midwife will tell you where to go for the blood draw. After that, your blood will be tested. The NIPT is free during pregnancy.



The NIPT for twins
Are you expecting twins, triplets or more? You can still choose to have the NIPT. Go to www.screening-cn.com/en/niptseo/how-does-the-nipt-work.



What is a chromosomal abnormality?



23 times 2 chromosomes



2 chromosomes

We have chromosomes in every cell in our body. These chromosomes are made up of DNA. Your DNA determines your physical characteristics: height, eye color, and how everything in your body works. Each cell has 23 sets of 2 chromosomes. Sometimes there is a problem when the chromosomes are formed. This is called a chromosomal abnormality, also known as a genetic disorder.

What types of chromosomal abnormalities are there?

There are different types of chromosomal abnormalities:



Extra chromosome: one too many



Extra part of a chromosome



Missing part of a chromosome

Extra chromosome: one too many

Down syndrome
A child born with Down syndrome will have it for the rest of their life. It will not go away. People with Down syndrome have intellectual disabilities, and you can see that they have it. Children with Down syndrome develop slower and less well than average. The average life expectancy for people with Down syndrome is 60 years old. They will need guidance and support from their families or others for their whole lives.





Edwards' syndrome and Patau's syndrome
These genetic disorders are very severe. Most children with Edwards' syndrome or Patau's syndrome do not survive to the end of the pregnancy, or die soon after birth.

Missing or extra part of a chromosome

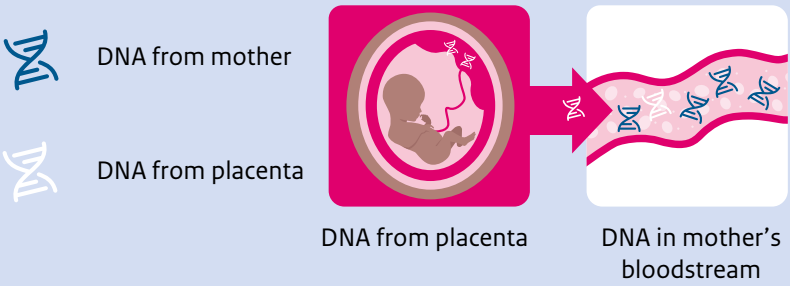
This is usually a serious genetic disorder. The child may have intellectual and physical disabilities.

Does the NIPT give clear results?

The results of the NIPT are not a 100% guarantee. Still, the results are usually reassuring: if the NIPT shows no indications of a chromosomal abnormality, your child is very unlikely to have one.

What if the results do show evidence of a chromosomal abnormality?
Then you can have more tests to confirm if the child does have a chromosomal abnormality. Even if the test does find evidence, it could come from the placenta, or (very rarely) from the mother. Your midwife can tell you more. It is up to you to decide whether you want to have follow-up tests. A chromosomal abnormality in a child always has serious consequences. But you cannot predict how serious it will be. It is different for every child.

Blood tests
The NIPT checks the DNA found in the blood taken from the pregnant mother. That blood mainly has the mother's DNA. But it also has some DNA from the placenta.



When do you get the test results?

Your midwife will usually give you the results within 14 days.

The results

These are the results that you could have:

1. There is no evidence of any chromosomal abnormality



About 993 in 1000 pregnant people get this result.

It is almost always accurate. It is very unlikely that you are carrying a child with a chromosomal abnormality. Fewer than 1 in 1000 pregnant people who get this result are carrying a child with a chromosomal abnormality.

No follow-up testing is needed.

2. There is evidence of a chromosomal abnormality

About 7 in 1000 pregnant people get this result.

You may be carrying a child with a chromosomal abnormality.
There are two options:

1. There is evidence of Down syndrome, Edwards' syndrome or Patau's syndrome.
2. There is evidence of some other chromosomal abnormality.

You will be invited for an appointment to discuss the results. You will get more information about the specific abnormality and about the options for follow-up testing.

3. The test did not work

Sometimes the test does not work. If that happens, you can choose to repeat the NIPT.

The 20-week ultrasound

In the 20-week ultrasound, an ultrasound technician uses a device to check if the unborn child has any physical abnormalities. The ultrasound technician also checks how much amniotic fluid is present and how the unborn child is growing.

Watch the video about the 20-week ultrasound



What happens during the ultrasound?

The 20-week ultrasound can only be done by specially trained ultrasound technicians. The ultrasound technician scans your unborn child by moving the ultrasound scanner over your belly. The ultrasound does not hurt at all and is not dangerous. Your unborn child will not feel anything. The ultrasound will take about 30 minutes. The 20-week ultrasound is free during pregnancy. Sometimes the ultrasound technician cannot see all the details clearly. For example, if there is excess weight or scar tissue in the way, or if the child is not positioned well.

Internal ultrasound (vaginal scan)
If it is hard to see all the details, the technician may suggest an internal ultrasound. That can be done right away. If you do not want an internal ultrasound, you can say no.



When can you have the 20-week ultrasound done?

You can have the 20-week ultrasound done in week 18+0 until week 21+0 of your pregnancy. That means it can be done from 18 weeks and 0 days until 21 weeks and 0 days of pregnancy. The ideal time for the ultrasound is in week 19 of your pregnancy: That means from 19 weeks and 0 days until 19 weeks and 6 days of pregnancy.

The ultrasound is not a guarantee

Did the ultrasound technician see something that needs to be checked? It is not always clear if it is a physical abnormality. Even if there is a possible abnormality, it is not clear how serious it is and what that means for your child. That is why follow-up testing is always offered.

The ultrasound technician cannot see all physical abnormalities on the 20-week ultrasound. That means that your unborn child may have a birth defect, even if the ultrasound scan does not show any physical abnormalities.

When do you get the test results?

The ultrasound technician will tell you the results right after the ultrasound scan.

What are the possible results of the 20-week ultrasound?

1. There is no evidence of abnormalities



About 950 in 1000 pregnant people get this result.

No follow-up testing is needed.

2. The ultrasound technician wants to repeat the scan

This means that the ultrasound technician could not see your child clearly. It does not mean that there are any problems with your child. The child may be lying in a position that is hard to see. If the ultrasound technician is not sure, you will be asked if you want another ultrasound. If you do, then the ultrasound will be repeated.

3. An abnormality was observed. Follow-up testing is needed to confirm

About 50 in 1000 pregnant people get this result.

This result means that the ultrasound technician saw something unusual. More tests are needed to find out exactly what your child has – and what that means for you and your child.

You can choose to have follow-up testing.
The decision is up to you.

4. An abnormality was observed. Follow-up testing is not needed

You will have another ultrasound later in your pregnancy. When that happens, the ultrasound technician checks if it went away.

Follow-up testing

Do you have the results of the NIPT or the 20-week ultrasound? And is there evidence that your child has a chromosomal or physical abnormality? If so, follow-up testing is needed to understand what it is. The ultrasound technician, midwife or doctor will tell you what options are available for follow-up testing and where you can go. It is up to you to decide whether you want to have follow-up tests.

What are your options for follow-up testing?

Which follow-up tests are offered depends on which test you had first and how far along you are in your pregnancy:

After the NIPT, follow-up testing could be:
- A comprehensive ultrasound
- Amniocentesis

After the 20-week ultrasound, follow-up testing could be:
- A comprehensive ultrasound
- A blood test
- Amniocentesis

In **amniocentesis**, the doctor removes a small amount of amniotic fluid and tests it. This can be done after week 16 of pregnancy.

The disadvantage is that there is a minor risk of miscarriage after amniocentesis. This happens in 1 out of 200 women

If you would like to know more about follow-up testing?

you will have an appointment to talk about it. You will not have to decide until after that. You may also decide that you do not want follow-up testing.

More information

Costs

The detailed talk and the tests are free. The government pays for the costs.

If there is evidence that your child has a chromosomal or physical abnormality, you can choose to have follow-up testing. These costs are paid by the Directorate for Youth and Care in the Caribbean Netherlands (ZJCN).

Data and privacy

What happens to my data?
If you choose to take part in prenatal screening, your care providers will keep your details about these tests in your medical records. This is required by law. Data and results are used to check whether care providers and the laboratory are doing their jobs properly. And they are used to estimate how many people choose to have prenatal screening during pregnancy.

Did you decide to have the NIPT?
After your blood is analyzed in the NIPT laboratory, some blood may be left over. The NIPT laboratory stores the blood and associated data in a well-secured system.

About this leaflet

This leaflet has been formulated with all due care. It is not possible to guarantee that it does not contain any errors.

More information

For more information about the NIPT and the 20-week ultrasound, go to www.screening-cn.com/en/niptseo.

Information in other languages

This information is available in Dutch / Papiamento / Spanish:

Deze informatie is ook beschikbaar in het Nederlands.

Bo por lesa e informashon aki den papiamentu.

Esta informacion esta disponible en Espanol.

Published by

National Institute for Public Health and the Environment, RIVM
www.rivm.nl/en

December 2025

016633