

# Information about the screening for *Down syndrome, Edwards' syndrome and Patau's syndrome*

Prenatal screening  
April 2017



## Brief summary of the prenatal screening for Down syndrome, Edwards' syndrome and Patau's syndrome

You have the opportunity to investigate (screen for) the risk of your child having Down syndrome, Edwards' syndrome or Patau's syndrome. People with Down syndrome have an intellectual disability as well. Their development cannot be predicted. They often suffer from health problems; in general, these can be treated well. In most cases, children with Edwards' syndrome or Patau's syndrome die prior to or during birth. They rarely live past the age of one. These children suffer from serious intellectual disability and serious physical abnormalities.

The screening: how does it work?

- During your first visit, your obstetrician or gynaecologist will ask you if you want to know more about the screening. If you want, you will be informed about the screening during an information session.
- You can prepare for this session by reading the information on the website [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) and by completing the *Bewust kiezen* (conscious decision) questionnaire.
- You can choose one of two different tests:
  - The combined test. This is a blood test for the mother, and a nuchal fold measurement for the foetus, using an ultrasound scan.
  - The NIPT (non-invasive prenatal test). This is a blood test for pregnant women.
- The NIPT can detect more cases of Down syndrome, Edwards' syndrome and Patau's syndrome, and is more accurate than the combined test (i.e. fewer pregnant women are unduly referred for follow-up examinations).
- After this session, you can decide if you want to participate in the screening. Participation in the screening is voluntary.
- The screening costs €168 (combined test for one child) or €175 (NIPT). You will be charged for these costs.
- The result of the screening may lead to difficult choices. Did you test positive, and are you perhaps carrying a child with Down syndrome, Edwards' syndrome or Patau's syndrome? In that case, you will be offered follow-up testing to make sure the result is correct. There is help available to make this choice.
- The result of the follow-up testing may again lead to difficult choices. There is also help available at this stage.

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# 1 Information in this brochure

You can choose to have your unborn child checked for a number of congenital disorders. This is called prenatal screening. You can select two tests: (1) The prenatal screening for Down syndrome, Edwards' syndrome and Patau's syndrome; and (2) testing for physical abnormalities (20-week anomaly scan). You determine if you want to undergo these tests. This brochure will tell you more about the prenatal screening for Down syndrome, Edwards' syndrome and Patau's syndrome.

## Prenatal screening for Down syndrome, Edwards' syndrome and Patau's syndrome

During your first visit, your obstetrician or gynaecologist will ask you if you want to know more about the screening for Down syndrome, Edwards' syndrome and Patau's syndrome. Would you like to know more? You can request a comprehensive information session about this subject. This is called a counselling session.

### Preparation for the session

During the session, you will receive a lot of information. We advise you to go to [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) prior to the session. This website contains a video that explains the screening process. This website also contains information and the questionnaire *Bewust kiezen* that will help you make a conscious decision. If you do not understand certain elements, you can ask questions during the session.

## Before you decide to take part in the screening

Suppose that during the pregnancy, you want to know if your child has a congenital disorder. What could the result mean to you?

1. *You become worried:* the result indicates that your child may suffer from a disorder. In most cases, further examination is needed to ascertain this. Would you want that?
2. *There may be difficult choices ahead:* the follow-up examination may conclude that your child has a disorder. You must consider what you want to do. Would you like to carry the pregnancy to full term, or would you like to terminate the pregnancy? Would you like to have this choice, or would you rather not have to think about this?
3. *You will be reassured:* the screening does not show any disorders. Or the screening has detected a disorder that will not pose a lot of problems in your child's daily life. Even if you tested negatively, your child could still have a disorder. The screening will not detect all disorders.

You decide if you want to take part in the prenatal screening, and how far you are willing to take the testing. You can withdraw from the test procedure at any moment.



# 2 The disorders

## What is Down syndrome?

Down syndrome is a congenital disorder that cannot be cured. People with Down syndrome have an intellectual disability. In some cases the intellectual disability is worse than in others. The severity of the disability cannot be predicted.

## People with Down syndrome have an additional chromosome

Chromosomes are part of all our body cells and contain our hereditary characteristics. Down syndrome is a chromosomal abnormality. In normal cases, people have two copies of each chromosome. A person with Down syndrome has not two, but three copies of a specific chromosome (chromosome 21) in each cell. Another name for Down syndrome is trisomy 21. Out of 10,000 children that are born, between 11 and 16 will have Down syndrome.

## Development

Children with Down syndrome develop more slowly and to a more limited extent than the average child. This varies from one child to another. It is not possible to predict development. By stimulating development from an early age, children can now develop better than they could in the past. Parents can use different support programmes. Young children can grow up and be part of the family. In most cases, they can go to regular day care facilities. In very rare cases, the child will need a special day care centre. Most children with Down syndrome will start at a regular school.



A small group receives special needs education. On average, children will learn better speaking and reading skills when they attend a regular school, and will develop better interaction skills. After primary school, most children will attend special needs secondary education. Some children will attend a day care centre. Some adolescents with Down syndrome will realise that they cannot live the same life their peers live. As a result of this realisation, adolescents with Down syndrome can be shy, insecure and withdrawn. That is why they can sometimes display other reactions than expected by the people around them.

Half of all adults with Down syndrome live at home until they are approximately thirty years old. Most people with Down syndrome will live independently in supported accommodation. Most people with Down syndrome live in small living groups. People with Down syndrome will require support and guidance throughout their lives. The level of guidance depends on the degree of their intellectual disability. The average life expectancy for people with Down syndrome is 60 years old.

### What does having a child with Down syndrome mean for the parents and siblings?

Research has shown that almost all parents state that they love their child with Down syndrome, and that they are proud of their child. Most parents (8 out of 10) feel that their child has given them a more positive outlook on life. This is also the case for most siblings. They have stated that they want to stay involved in their adult sibling's life later on. Some families do encounter problems, and find it a strain on the family.

All scientific articles about this subject are available on [www.downsyndroom.nl/home/levensloop/kwaliteit-van-leven/](http://www.downsyndroom.nl/home/levensloop/kwaliteit-van-leven/).

## Health

The risk of a child with Down syndrome dying during pregnancy is higher than average. Almost half of all children with Down syndrome are born with a heart defect. In most cases, this can be treated with surgery. Usually that means that the child will not have any problems after that. Children with Down syndrome can also suffer from a gastrointestinal abnormality. This can also be treated with surgery. Children with Down syndrome have a higher risk of respiratory disorders, hearing, eye, and speech problems, and decreased immunity against infections. The seriousness of the health problems varies per person. Adults with Down syndrome are more likely to develop Alzheimer's disease, and at a younger age, than average.

## Support

Children and young people with Down syndrome, and their parents, can seek assistance from paediatricians, Down syndrome clinics, or Down syndrome teams. Down syndrome teams are composed of various professionals, such as a paediatrician, a speech therapist, a physiotherapist and a social worker. Adults with Down syndrome can seek assistance from their family doctor, Down syndrome clinic, or Down syndrome team. Healthcare insurance will cover the costs for medical care, including any resources, for children with Down syndrome. Parents can also request a number of financial aid arrangements.



## What is Edwards' syndrome?

Edwards' syndrome is a very serious congenital disorder. A child with Edwards' syndrome has not two, but three chromosome 18 copies in each cell. Another name for Edwards' syndrome is trisomy 18. This condition is much rarer than Down syndrome. 1 out of 10,000 children that are born will suffer from Edwards' syndrome.

The majority of children with Edwards' syndrome will die during pregnancy or shortly after birth. Children are often born with retarded growth. Children with Edwards' syndrome who are born alive, will have a low birth weight. Their health is very fragile and they usually die during their first year. Children with Edwards' syndrome have a severe intellectual disability. Approximately 9 out of 10 children will have a serious congenital heart defect. In many cases, other organs, such as the kidneys and intestines, are also affected. Children can also suffer from abdominal wall defect and oesophageal atresia. The child can have a small face and a large cranium. The health problems are always serious. The nature and severity of the problems can vary per child.

## What is Patau's syndrome?

Patau's syndrome is a very serious congenital disorder. A child with Patau's syndrome has not two, but three chromosome 13 copies in each cell. Another name for Patau's syndrome is trisomy 13. This condition is much rarer than Down syndrome. 1 out of 10,000 children that are born will suffer from Patau's syndrome.

The majority of children with Patau's syndrome will die during pregnancy or shortly after birth. Children are often born with retarded growth. Children with Patau's syndrome who are born alive, will have a low birth weight. Their health is very fragile and they usually die during their first year. Children with Patau's syndrome have a severe intellectual disability. There is usually a developmental abnormality of the brain and heart. In some cases, children also have kidney disorders and gastrointestinal tract abnormalities. The child may also have extra fingers or toes. These children may also have facial abnormalities, such as a cleft lip-jaw-palate. The health problems are always serious. The nature and severity of the problems can vary per child.

# 3 The screening

Have you decided that you want to be tested for Down syndrome, Edwards' syndrome and Patau's syndrome? In that case, you can select one of two tests:

1. the combined test
2. the NIPT

## The combined test

The combined tests consists of a combination of two tests:

1. A blood test of the blood of the mother in the period between the 9th and 14th week of the pregnancy. The blood is tested in a laboratory.
2. An ultrasound scan measuring the nuchal fold of the foetus in the period between the 11th and 14th week of the pregnancy. All children have a thin layer of fluid in the nuchal fold under the skin in their neck. The thicker the skin fold in the neck, the greater the likelihood that the child will have Down syndrome, Edwards' syndrome or Patau's syndrome.

## The result of the combined test

The combined test calculates the *risk* of your child having Down syndrome, Edwards' syndrome or Patau's syndrome. The test will not provide any certainty. In case of an increased risk of a child with Down syndrome, Edwards' syndrome or Patau's syndrome, you can decide whether you want to have additional follow-up testing done (see chapter 4 about 'Follow-up testing'). This follow-up test will clearly show whether your child has any of these syndromes or not.

*“You have an increased risk of a child with Down syndrome, Edwards' syndrome or Patau's syndrome.”*

This means that there is a risk of 1 in 200 or more that you are carrying a child with Down syndrome, Edwards' syndrome or Patau's syndrome. A risk of 1 in 200 means that out of every 200 pregnant women, 1 woman is pregnant with a child with Down syndrome, Edwards' syndrome or Patau's syndrome. The other 199 women are not expecting a child with Down syndrome, Edwards' syndrome or Patau's syndrome. An increased risk is not the same as a high or significant risk. For more certainty about your situation, you can decide to participate in the follow-up test.

*“You do not have an increased risk of a child with Down syndrome, Edwards' syndrome or Patau's syndrome.”*

This means that the risk of having a child with Down syndrome, Edwards' syndrome or Patau's syndrome is less than 1 in 200. You do not need follow-up testing. The result of the combined test will indicate the risk, but it is not a guarantee. There is still a small risk of your child having Down syndrome, Edwards' syndrome or Patau's syndrome. Go to [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) for more information about the combined test and an explanation about the risks.

### **Additional findings of the combined tests**

If the skin fold is 3.5mm or more, you will always be offered additional ultrasound testing. A thickened skin fold is not only an indication of Down syndrome, Edwards' syndrome and Patau's syndrome, but may also indicate other chromosomal abnormalities and physical disorders in the child, such as heart defects. In some cases, there is no clear cause for the thickened skin fold, and the child is born without any disorders.

In some cases, the ultrasound technician will discover other disorders during the nuchal fold test (additional findings). These are serious abnormalities, such as missing limbs or brains.

If you are taking part in the combined test, you will always be informed of any additional findings.

### **The NIPT**

The NIPT tests the blood of the pregnant woman. The blood is drawn and tested. The laboratory tests the DNA in the blood for chromosomal abnormalities, allowing the determination of whether the child has Down syndrome, Edwards' syndrome or Patau's syndrome. The blood of the pregnant woman contains placenta DNA and DNA from the mother. The placenta DNA is almost always the same as the DNA of the child.

The NIPT can be performed from the 11th week of the pregnancy.

## Scientific study

From 1 April, 2017, all pregnant women in the Netherlands can choose the NIPT, but only if they participate in a scientific study (TRIDENT-2). Why a study? Other countries already have more experience in the use of the NIPT. Their results have shown that the NIPT is a very reliable test. The minister now wants to investigate if the test is also successful in the Netherlands, and what women think about the NIPT. If you select the NIPT, you are giving researchers permission to use your data. You sign a consent form for this part of the test.

The website [www.meerovernipt.nl](http://www.meerovernipt.nl) tells you more about the scientific study and what your data will be used for.

## Additional findings of the NIPT

The laboratory can also detect other chromosomal abnormalities, besides Down syndrome, Edwards' syndrome or Patau's syndrome. These are detected in the foetus, in the placenta and, very rarely, in the pregnant woman herself. These are additional findings. You decide whether you want to be informed about additional findings.

There are different types of additional findings, ranging from very serious to moderately serious. To determine the type of additional findings, an amniocentesis or chorionic villus sampling is usually required.

Approximately 4 out of 1000 pregnant women who have decided to take part in the NIPT will be informed about an additional finding.

Go to [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) for more information about the NIPT and additional findings.

If you select the NIPT, you can decide afterwards if you want to be informed about any additional findings. There are two options:

1. You only want to have your unborn child screened for Down syndrome, Edwards' syndrome and Patau's syndrome. The laboratory will not know if there are any additional findings.

or

2. You want to have your child screened for Down syndrome, Edwards' syndrome and Patau's syndrome, and you want to know if the lab has detected any additional findings.

## The result of the NIPT

### *“The result is non-abnormal.”*

This result is almost always correct. The risk of carrying a child with Down syndrome, Edwards' syndrome or Patau's syndrome is very small. You do not require any follow-up testing. Your gynaecologist or obstetrician will inform you about the result.

### *“You have an abnormal result, and could perhaps be carrying a child with Down syndrome, Edwards' syndrome or Patau's syndrome.”*

The gynaecologist or obstetrician will inform you of this result. What does this result mean? Here are some examples:

- In 75 out of 100 women that have an abnormal result stating that they might be pregnant with a child with Down syndrome, this is in fact true; therefore, 25 women are not carrying a child with Down syndrome.
- In 24 out of 100 women that have an abnormal result stating that they might be pregnant with a child with Edwards' syndrome, this is in fact true; therefore, 76 women are not carrying a child with Edwards' syndrome.
- In 23 out of 100 women that have an abnormal result stating that they might be pregnant with a child with Patau's syndrome, this is in fact true; therefore, 77 women are not carrying a child with Patau's syndrome.

In case of an abnormal result, there is a chance that the child does not have the disorder. You can only be certain if you have a chorionic villus sampling test or amniocentesis. Are you considering the termination of the pregnancy? In that case, you must first undergo a follow-up test to confirm the result.

### *“We have detected an additional finding.”*

Someone will call you and explain to you what has been detected, and what this might mean for your child or yourself. You will be invited to a session at the outpatient clinic for clinical genetics at an academic hospital. During this session, they will inform you about the additional finding and the options (see also page 22). Follow-up testing is required to confirm the result.

### *“We have not detected any additional findings.”*

If the results do not mention any additional findings, this means there are no additional findings.

## The combined test and NIPT compared

	Combined test	NIPT
<b>How does the testing work?</b>	Blood testing for pregnant women and nuchal fold measurements with an ultrasound scan for the foetus.	Blood testing for pregnant women.
<b>Is this test a scientific study?</b>	No, this test has been around for a while in the Netherlands.	Yes, this is a new test in the Netherlands. You must give your consent for your data to be used for the scientific study. See also <a href="http://www.meerovernipt.nl">www.meerovernipt.nl</a> .
<b>When can I take the test?</b>	Blood test in the period between weeks 9 and 14 of the pregnancy.	From the 11th week of the pregnancy.
<b>How long does it take before I get the result?</b>	This depends on the centre where the ultrasound scan is performed. If the blood was taken one or two weeks prior to the scan, you will usually get the result on the day of the scan. If the blood was taken later, you will usually get the result a couple of days after the scan.	Within 10 working days.
<b>Will the test detect all cases of Down syndrome, Edwards' syndrome and Patau's syndrome?</b>	No. The test will detect approximately <ul style="list-style-type: none"> <li>• 85 of 100 cases of Down syndrome,</li> <li>• 77 of 100 cases of Edwards' syndrome,</li> <li>• 65 of 100 cases of Patau's syndrome.</li> </ul>	No. The test will detect approximately <ul style="list-style-type: none"> <li>• 96 of 100 cases of Down syndrome,</li> <li>• 87 of 100 cases of Edwards' syndrome,</li> <li>• 78 of 100 cases of Patau's syndrome.</li> </ul>

	<b>Combined test</b>	<b>NIPT</b>
<b>Does the test give any guarantees?</b>	<p>No. The test indicates the chances of a child with a disorder. The result (1 in x cases) will indicate the chance that the result is right.</p> <ul style="list-style-type: none"> <li>• Is the chance smaller than 1 in 200 (for instance 1 in 1000)? There is no increased risk. You do not require any follow-up testing.</li> <li>• Is the chance 1 or 200 or higher (for instance 1 in 50)? There is an increased risk. You will be offered follow-up testing to make sure the result is right.</li> </ul>	<p>No.</p> <ul style="list-style-type: none"> <li>• Is the result non-abnormal? This is almost always correct. You do not require any follow-up testing.</li> <li>• Do you have an abnormal result? Approximately 75 out of 100 women with this result are actually carrying a child with Down syndrome. For Edwards' and Patau's syndrome, these numbers are 24 and 23 out of 100, respectively, for women with an abnormal result. You will be offered follow-up testing to make sure the result is right.</li> </ul>
<b>Can the test also detect unexpected additional findings?</b>	<p>The skin fold measurement may also detect other, often serious disorders. These are serious abnormalities, such as missing limbs or brains, or an abdominal wall defect.</p>	<p>The NIPT may indicate chromosomal abnormalities in the child, in the placenta and, in very rare cases, in the mother. The abnormalities may vary from very severe to moderately severe.</p>
<b>Do I get to choose whether I want to find out about the additional findings?</b>	<p>Any additional findings are always reported to you.</p>	<p>You can choose not to be informed about any additional findings.</p>
<b>What does the test cost?</b>	<p>€168 (for single foetus)</p>	<p>€175</p>

## A list of the differences

You want to screen for Down syndrome, Edwards' syndrome and Patau's syndrome, but you find it difficult to select one of the two tests? Use the table on pages 14 and 15 to compare the tests. Studies have shown that the NIPT detects more cases of Down syndrome, Edwards' syndrome and Patau's syndrome than the combined test, and is also more accurate (which means that fewer pregnant women are referred for follow-up testing). You will find more information about the tests and a more comprehensive comparison table at [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl). Your obstetrician or gynaecologist can also tell you more about the tests.

## Screening in twins

Are you expecting twins? In that case, you can choose the combined test, and in some cases the NIPT. You can get more information about this from your obstetrician or gynaecologist.

### *If you choose the combined test*

You will receive separate results for each child. If one or both children has an increased risk of Down syndrome, Edwards' syndrome or Patau's syndrome, you will be offered a follow-up test.

### *If you choose the NIPT*

If you are carrying twins, the NIPT is not always an option. Your obstetrician or gynaecologist can tell you more about this.

## Do you have a medical indication?

You have a medical indication, for instance if you have already had a child with Down syndrome, Edwards' syndrome or Patau's syndrome. Are you considering prenatal screening? You will be invited to a session at a Prenatal Diagnostics Centre.

If you decide that you want your child to be screened for Down syndrome, Edwards' syndrome and Patau's syndrome, you can discuss the best option for your situation.



## What role does the age of the mother play?

Older pregnant women have a higher risk of a child with Down syndrome than younger women. This is also the case for Edwards' syndrome or Patau's syndrome.

The age of the pregnant woman	Risk of child with Down syndrome at the time of the screening
20 – 25 years	11 to 13 out of 10,000
26 – 30 years	14 to 19 out of 10,000
31 – 35 years	20 to 45 out of 10,000
36 – 40 years	60 to 155 out of 10,000
41 – 45 years	200 to 615 out of 10,000

### *Explanation table*

If 10,000 25-year-old women are pregnant, 13 of them are carrying a child with Down syndrome. This means that 9,987 women are pregnant with a child without Down syndrome. If 10,000 41-year-old women are pregnant, 200 of them are carrying a child with Down syndrome. This means that 9,800 women are pregnant with a child without Down syndrome.

You can also go to [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) for more information.

# 4 Follow-up examination

Have you tested positive? You can decide not to take part in any follow-up tests, and to continue your pregnancy. You are not obligated to undergo follow-up tests. Would you like to know for certain if your child is suffering from a disorder? In that case, you can decide to undergo follow-up testing. If you are considering a termination of your pregnancy, you will be required to undergo a follow-up test. If you think you might want to undergo follow-up testing, you can discuss this at the Prenatal Diagnostics Centre. After that, you can decide what you want to do.

## Was the first test a combined test?

You can decide to undergo follow-up testing if there is an increased risk of Down syndrome, Edwards' syndrome or Patau's syndrome. An increased risk is a risk of 1 in 200 or more, for instance 1 in 150, or 1 in 100, or 1 in 20. For more explanation about an increased risk please go to [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) and [www.erfelijkheid.nl](http://www.erfelijkheid.nl).



If the combined test has shown you are at an increased risk, these follow-up test options are available to you:

1. You can select the NIPT: The advantage of the NIPT is that there is no risk of miscarriage. On the other hand, the NIPT is not 100% guaranteed. If the NIPT shows no abnormal results, you are very likely not carrying a child with Down syndrome, Edwards' syndrome or Patau's syndrome. You will not be offered an amniocentesis or chorionic villus sampling. If the NIPT does indicate an abnormal result, you can still decide to undergo an amniocentesis or chorionic villus sampling to be sure. You cannot opt for the NIPT if you are carrying non-identical twins or if the ultrasound scan has indicated abnormalities such as a thickened nuchal fold.
2. In that case, you can opt directly for an amniocentesis or chorionic villus sampling.

## Was the first test a NIPT?

If your NIPT result was abnormal, you can decide to undergo follow-up testing. The follow-up testing is a chorionic villus sampling test, or an amniocentesis.

## Amniocentesis and chorionic villus sampling

The chorionic villus sampling can be performed from the 11th week of the pregnancy. The specialist will take a sample of the placenta tissue for testing. The amniocentesis can be performed from the 15th week of the pregnancy. During an amniocentesis, the specialist will take a sample of the amniotic fluid for testing. An amniocentesis or chorionic villus sampling will give you guaranteed results about your child having Down syndrome, Edwards' syndrome or Patau's syndrome. The disadvantage of these tests is that there is an increased risk of miscarriage as a result of the test. This occurs in 2 out of 1000 women who undergo this test; so 998 women will not have a miscarriage as a result of this test.

Would you like more information about the chorionic villus sampling or the amniocentesis? Please go to [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) and [www.erfelijkheid.nl](http://www.erfelijkheid.nl).

# 5 Conscious decision

You decide if you want to be tested for Down syndrome, Edwards' syndrome and Patau's syndrome. What are your considerations? You can consider the following elements:

- How much do you want to know about your child before it is born?
- Suppose you test positive, and your child may have a disorder. Would you want to have follow-up testing done to make sure? You can also do nothing and carry the foetus to term. You decide what you want to do.
- What is your opinion about chorionic villus sampling or amniocentesis and its increased risk of a miscarriage?
- If the follow-up test shows that your child indeed suffers from a disorder, how can you prepare for this?
- What are your thoughts about life with a child with Down syndrome, Edwards' syndrome or Patau's syndrome?
- What is your opinion about potentially terminating a pregnancy if the child has a disorder?

The follow-up test may show that you are carrying a child with Down syndrome, Edwards' syndrome or Patau's syndrome. It is also possible that you are carrying a child with a different chromosomal abnormality. This may mean that you have some difficult decisions to make. Discuss this with your partner, with your obstetrician, family doctor, gynaecologist or other people who play an important role in your life. If you decide that you want to terminate your pregnancy, you can do so up to the 24th week of your pregnancy.

Regardless of the decision you make, your obstetric care provider will always offer you guidance.

## Help in choosing

At [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl) you will find the questionnaire *Bewust kiezen* (conscious decision). This can help you organise your thoughts about the screening. The site also contains tips on how to discuss this subject with your partner or others. The meeting with your obstetrician or gynaecologist is also intended to help you make a conscious decision.

# 6 What else do you need to know?

## Comprehensive information session (counselling)

Are you considering to have your child screened for Down syndrome, Edwards' syndrome and Patau's syndrome? You will first be meeting with your obstetrician or gynaecologist to discuss this matter extensively. They will answer the following questions:

What are Down syndrome, Edwards' syndrome and Patau's syndrome?

- What are the available tests?
- What are the advantages and disadvantages of these tests?
- How long does it take before you know more?
- What does the screening cost?
- What follow-up testing is available in case you test positive?

If you have any questions, please ask these during the session. Do you have any doubt about the screening after the session? You can discuss this again with your obstetrician or gynaecologist. A second meeting could offer you more clarity. After the extensive session (counselling), you decide if you want to take part in the screening for Down syndrome, Edwards' syndrome and Patau's syndrome.



## Preparing for the session

You can prepare yourself by reading the information on [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl). During the session, you can ask questions, allowing your obstetrician or gynaecologist to help you make a decision.

## When and how do you get the results?

The timing of the results depends on the test, and varies per obstetrician, gynaecologist and/or hospital. You will be informed about this prior to the test. See also the table on page 14. Have you also decided to be informed about any additional findings of the NIPT, and has the laboratory detected any additional findings? In that case, you will hear the result from your obstetric care provider. In some cases, you will be called by an expert at one of the Prenatal Diagnostics Centres or an outpatient clinic for clinical genetics at one of the academic hospitals.

## Costs and reimbursements of the prenatal screening

### *Costs of a comprehensive information session (counselling)*

Your healthcare insurance will pay for the costs of the comprehensive information session (counselling) with your obstetrician or gynaecologist about the possible screening for Down syndrome, Edwards' syndrome and Patau's syndrome. This has no consequences for the excess risk part of your healthcare insurance. You do not have to pay anything.

### *Costs of the screening*

You will have to pay for the tests. The combined test costs €168 (single foetus), and for the NIPT you will pay a contribution of approximately €175. You can ask your healthcare insurer whether the tests are covered by your additional insurance.

### *Costs of screening in case of medical indication*

Do you have a medical indication? In that case, you will be invited to a session at a Prenatal Diagnostics Centre. Together with an expert, you will decide what the best test is in your case. Your healthcare insurer will pay the costs of the session and the test, but this may have to be covered by your health insurance excess. You can ask your healthcare insurer about this.

### *Costs and reimbursements for follow-up testing*

If you tested positive in the combined test or the NIPT, you can take part in follow-up testing (see page 18). The costs of this testing are covered by your basic healthcare insurance. This may have to be charged to your health insurance excess. You can ask your healthcare insurer about this.

The expert at the Prenatal Diagnostics Centre will be able to tell you more about this.

### *Costs and reimbursements may change*

The costs and reimbursements described above may change. For current information about the costs: **[www.onderzoekvanmijnongeborenkind.nl/kosten](http://www.onderzoekvanmijnongeborenkind.nl/kosten)**.

You should also always check the conditions of your health insurance.

## **Agreement required**

The comprehensive session (counselling) and the combined test should only be performed by a care provider who has signed an agreement with the Regional Centre for Prenatal Screening. The comprehensive session (counselling) will only be reimbursed if the care provider has such an agreement. We would advise you to check this in advance with your obstetrician, family doctor or gynaecologist.

At **[www.onderzoekvanmijnongeborenkind.nl/kosten](http://www.onderzoekvanmijnongeborenkind.nl/kosten)** you can see the contracted obstetricians or gynaecologists in your area. It is also wise to check if the healthcare insurer has a contract with the care provider. You can ask your healthcare insurer about this.

# 7 Additional information

## Internet

The information from this brochure is also published on [www.onderzoekvanmijnongeborenkind.nl](http://www.onderzoekvanmijnongeborenkind.nl). Do you find it difficult to decide if you want to be tested for Down syndrome, Edwards' syndrome and Patau's syndrome? Perhaps the *Bewust kiezen* (conscious decision) questionnaire on the site can help you. The site also contains tips on how to discuss this subject with your partner or others.

Other websites with information about prenatal screening:

[www.erfelijkheid.nl](http://www.erfelijkheid.nl)

[www.deverloeskundige.nl](http://www.deverloeskundige.nl)

[www.thuisarts.nl](http://www.thuisarts.nl)

[www.meerovernipt.nl](http://www.meerovernipt.nl)

## Information sheets

Would you like to know more about the disorders in this brochure?

Ask your obstetrician or gynaecologist about the information sheets.

There are information sheets about:

- Down syndrome (trisomy 21)
- Trisomy 18 (Edwards' syndrome)
- Trisomy 13 (Patau's syndrome)
- Spina bifida and anencephaly.

You can also download these information sheets at

[www.rivm.nl/down-edwards-patau/informatieblad](http://www.rivm.nl/down-edwards-patau/informatieblad) and

[www.rivm.nl/zowekenecho/informatieblad](http://www.rivm.nl/zowekenecho/informatieblad).



## Brochures about other prenatal tests

There is a separate information brochure about the 20-week anomaly scan. This test is also part of the prenatal screening. This brochure is located at [www.rivm.nl/zowekenecho/folder](http://www.rivm.nl/zowekenecho/folder). The leaflet *Zwanger! (Pregnant!)* contains general information about the pregnancy. This leaflet also contains information about the blood test that you will undergo before you are 12 weeks pregnant. This blood test will determine, among other things, your blood type, and detect any infectious diseases. This leaflet can be found on [www.rivm.nl/folderzwanger!](http://www.rivm.nl/folderzwanger!) You can also ask your obstetrician, family doctor or gynaecologist about the leaflets.



## Organisations and addresses

### **The Dutch Down Syndrome Foundation**

This is a parents' association that strives to promote the interests of people with Down syndrome, and those of their parents. The foundation can provide you with further details on Down syndrome. The foundation also supports pregnant couples who want to determine if they can handle life with a child with Down syndrome. They will be informed about life with Down syndrome, allowing them to make a suitable decision. The foundation also provides support to the parents of newborn children with Down syndrome.

**[www.downsyndroom.nl](http://www.downsyndroom.nl)**

Email: [helpdesk@downsyndroom.nl](mailto:helpdesk@downsyndroom.nl)

Telephone: +31 (0)522 - 281 337

### **The Erfocentrum**

The Erfocentrum is the Dutch information centre on genetics.

**[www.erfocentrum.nl](http://www.erfocentrum.nl), [www.erfelijkheid.nl](http://www.erfelijkheid.nl), [www.zwangerwijzer.nl](http://www.zwangerwijzer.nl)**

Email Erfolijn: [erfolijn@erfocentrum.nl](mailto:erfolijn@erfocentrum.nl)

Erfolijn: +31 (0)33 - 303 2110

### **VSOP**

The Dutch Genetic Alliance (VSOP) is involved in genetic issues. VSOP is an umbrella organisation of approximately 60 patient organisations, most of which focus on genetic, congenital or rare disorders. For over 30 years, VSOP has been representing their collective interests in the field of genetic issues, ethics, pregnancy, biomedical research and care for rare disorders.

**[www.vsop.nl](http://www.vsop.nl)**

Telephone: +31 (0)35 - 603 4040

### **Association of VG networks**

The Association of VG networks is a networking group for individuals with very rare syndromes associated with an intellectual disability and/or learning difficulties, and their parents.

**[www.vgnetwerken.nl](http://www.vgnetwerken.nl)**

Email: [info@vgnetwerken.nl](mailto:info@vgnetwerken.nl)

Telephone: +31 (0)30 - 720 0030

### **Platform ZON**

This is a patient organisation for parents of children with very rare or unknown disorders, including chromosomal abnormalities such as Edwards' syndrome and Patau's syndrome.

**[www.ziekteonbekend.nl](http://www.ziekteonbekend.nl)**

### **RIVM**

The RIVM coordinates screening programmes for Down syndrome, Edwards' syndrome and Patau's syndrome, and physical defects at the request of the Ministry of Health, Welfare and Sport, in cooperation with the various medical professional associations. Further details: **[www.rivm.nl/down-edwards-patau-seo](http://www.rivm.nl/down-edwards-patau-seo)** under Organisatie (organisation).

### **Regional Centres for Prenatal Screening**

The eight regional centres are all licensed to organise the screenings described above. They maintain contractual agreements with the screeners and are responsible for regional quality assurance. For more information on these regional centres, visit: **[www.rivm.nl/down-edwards-patau-seo](http://www.rivm.nl/down-edwards-patau-seo)** under Organisatie (organisation).

# 8 The use of your data

If you decide to participate in the screening for Down syndrome, Edwards' syndrome and Patau's syndrome, your data will be used. Without this data, it would not be possible to make an accurate diagnosis, to offer effective treatment or to safeguard the quality of care. This data is recorded in your own medical file, and in the Peridos database. This system is used by all health care providers who are involved in prenatal screening in the Netherlands. However, only those healthcare providers who are involved in your screening will be able to access your details. The system includes effective safeguards to protect your privacy.

If necessary, the regional centre can also access the data held in Peridos. The regional centre coordinates the screening programme and monitors compliance with quality standards by all the healthcare providers involved. To this end, it has obtained a permit from the Ministry of Health, Welfare and Sport (VWS). The screening is required to meet national quality standards. The screening must meet the national quality standards.

One of the ways in which the regional centre monitors quality involves the use of data stored in Peridos. The healthcare providers themselves also engage in quality control. To this end, they occasionally need to compare data. Your healthcare provider will be able to provide you with more details about the protection of your information. Would you prefer your personal information to be deleted from Peridos after the screening? If so, tell your obstetric healthcare provider.

## Scientific research

Aside from your healthcare providers and the regional centre, no-one can access your personal information. Information that is used for statistical purposes (to find out how many pregnant women make use of prenatal screening, for example) is entirely anonymous. This means that there is no way in which this information could be traced back to you – not even by those who produce the statistics in question.

Prenatal screening must be continually improved, so scientific research is needed. In the course of such research, your privacy will be protected.

Researchers will not be given access to your name or address. If your data are nevertheless required for the purpose of scientific research, those involved will always ask for your permission. Whatever your decision, this will not affect the way you are treated before, during, or after the screening.

More information about these subjects is located at [www.onderzoekvanmijnongeborenkind.nl/privacy](http://www.onderzoekvanmijnongeborenkind.nl/privacy)

## TRIDENT-1 and TRIDENT-2 study

If you selected the NIPT, you are also participating in a scientific study. Your data will be used. Go to [www.meerovernipt.nl](http://www.meerovernipt.nl) for further details.

## English

This brochure is designed to inform you (and your partner) about prenatal screening for Down syndrome, Edwards' syndrome and Patau's syndrome. The English brochure text is available on [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder).

## Deutsch

Diese Broschüre bietet Ihnen (und Ihrem Partner) Informationen über das pränatale Screening auf Down-, Edwards- und Patau-Syndrom. Sie finden den deutschen Text der Broschüre auf der Internetseite [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder).

## Français

Dans cette brochure, vous (votre conjoint ou compagnon) trouverez des informations sur le dépistage prénatal du syndrome de Down, d'Edwards et de Patau. Vous trouverez la version française de cette brochure sur [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder).

## Español

Este folleto ofrece información a usted y a su pareja, sobre el screening prenatal del síndrome de Down, Edwards y Patau. Encontrará el texto en español de este folleto en [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder).

## Polski

Niniejszą broszurę opracowano w celu poinformowania Ciebie oraz Twojego partnera/Twojej partnerki o przesiewowym badaniu w kierunku zespołu Downa, zespołu Edwardsa i zespołu Patau. Broszura w języku polskim jest dostępna na stronie [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder).

## Português

Esta brochura proporciona a si (e ao seu marido ou companheiro) informação sobre o exame pré-natal para deteção das síndromes de Down, Edwards e Patau. O texto desta brochura em português encontra-se em [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder).

## Papiamentu

Den e foyeto aki abo (i bo partner) ta haña informashon tokante screening prenatal di e síndrome di Down, Edwards i Patau. Bo ta haña e kontenido di e foyeto aki na Papiamentu na [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder).

## Türkçe

Bu broşür, Down, Edwards ve Patau sendromu için doğum öncesinde uygulanan tarama testi hakkında size (ve eşinize) bilgi vermek amacıyla hazırlanmıştır. Türkçe metne [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder) adresinde bulabilirsiniz.

عربي

تم إعداد هذا الكتيب لتعريفك (وتعريف شريكك) على الفحص السابق للولادة للكشف عن متلازمة داون وإدوارد وباتو. يمكن العثور على نص هذا الكتيب باللغة العربية على الموقع التالي [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder)

本信息小册是为您（和您的伴侣）提供关于唐氏、爱德华、帕陶综合症产前筛查的信息。本信息小册的中文版可以在 [www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder) 网站获取。

## Publishing details

The contents of this brochure were developed by a working group. This working group includes the organisations representing family doctors (NHG), obstetricians (KNOV), gynaecologists (NVOG), regional prenatal screening centres, ultrasound operators (BEN), paediatricians (NVK), clinical geneticists (VKGN), the Erfocentrum, the Dutch Genetic Alliance (VSOP), and the National Institute for Public Health and the Environment (RIVM).

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*This brochure outlines the current situation, based on the most recent available information. The parties responsible for drawing up this brochure accept no liability for any errors or inaccuracies. For a personal consultation, please contact your obstetrician, family doctor or gynaecologist.*

You can also access this brochure via **[www.rivm.nl/down-edwards-patau/folder](http://www.rivm.nl/down-edwards-patau/folder)**.

Obstetricians, gynaecologists, family doctors, ultrasound operators and other obstetric care providers can order additional copies of this brochure via the website **[www.rivm.nl/pns-folders](http://www.rivm.nl/pns-folders)**.

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National Institute for Public Health  
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*Ministry of Health, Welfare and Sport*



NEDERLANDSE VERENIGING VOOR  
OBSTETRIE & GYNAECOLOGIE



KONINKLIJKE NEDERLANDSE  
ORGANISATIE VAN VERLOSKUNDIGEN



BEROEPSVERENIGING  
ECHOSCOPISTEN  
NEDERLAND

VOOR VERLOSKUNDE EN GYNAECOLOGIE



VOOR ZELDZAME EN GENETISCHE AANDOENINGEN



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VERENIGING  
KLINISCHE GENETICA  
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8 REGIONALE  
CENTRA VOOR  
PRENATALE  
SCREENING