

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

Prenatal screening

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1 What can you get tested?

During pregnancy, you can have screening tests to find out whether your unborn baby has a congenital disorder or any physical abnormalities. This is called prenatal screening. There are two screening tests:

1. the prenatal screening for Down syndrome, Edwards' syndrome and Patau's syndrome, and
2. screening for physical abnormalities (anomaly scan).

It's entirely up to you to decide whether or not you want to take these screening tests. This leaflet contains further details about prenatal screening for Down syndrome, Edwards' syndrome and Patau's syndrome.

There is also a leaflet about the anomaly scan.

You can have tests (screening) to find out whether you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome. This screening test takes place in early pregnancy. During your first visit, your obstetric care provider will ask you if you would like to know more about screening for Down syndrome, Edwards' syndrome and Patau's syndrome.

The obstetric care provider is often your midwife, but can also be another care provider, like a gynecologist or a nurse.

What if you don't want any information about this screening? In that case, your obstetric care provider will not tell you anything about the screening. This means you have decided not to have your baby screened for Down syndrome, Edwards' syndrome and Patau's syndrome.

Would you like to know more? If so, then you will have an in-depth discussion about prenatal screening. This is called a counselling session. After this session, you are free to choose whether or not you want to take this screening test. During this session, your obstetric care provider will also discuss the anomaly scan.

Where can you find more information?

You will be given a lot of information during the counselling session. We recommend that you first take a look at www.onderzoekvanmijnongeborenkind.nl.

If there is anything you don't understand, please feel free to ask about it during the session.

This website features a video clip that explains the screening.

2 The disorders

What is Down syndrome?

Babies with Down syndrome are born with that disorder. They cannot recover from this disorder. People with Down syndrome have an intellectual disability and, in many cases, they also look different. The severity of the disability cannot be predicted.

People with Down syndrome have an extra chromosome.

Chromosomes are found in every cell in our body. They contain our hereditary characteristics. Down syndrome is a chromosomal abnormality (see below).

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 impossible to predict how a child will develop. It is good to stimulate such children, starting at birth. This will often improve its development. A variety of programmes that help such children to develop are now available to parents.

People with Down syndrome, Edwards' syndrome or Patau's syndrome have an extra chromosome

Chromosomes are found in every cell in our body. Chromosomes are made up of DNA. That DNA determines what our body looks like, and how everything in it works. Every cell contains 23 pairs of chromosomes. Someone with Down syndrome, Edwards' syndrome or Patau's syndrome has an extra chromosome in every cell.

- In someone with Down syndrome, each cell contains three copies of one specific chromosome (chromosome 21, to be exact), instead of the usual two. Another name for Down syndrome is trisomy 21.
- In a child with Edwards' syndrome, each cell contains three copies of one specific chromosome (chromosome 18, to be exact), instead of the usual two. Another name for Edwards' syndrome is trisomy 18.
- In a child with Patau's syndrome, each cell contains three copies of one specific chromosome (chromosome 13, to be exact), instead of the usual two. Another name for Patau's syndrome is trisomy 13.

If you want to know what the likelihood is that a baby will have Down syndrome, Edwards' syndrome and Patau's syndrome, take a look at www.onderzoekvanmijnongeborenkind.nl.

Young children just grow up as members of the family. They can usually go to normal childcare facilities. In a very few cases, they need to go to a special day care centre. Most children with Down syndrome start their school career at normal primary schools. A small number receive special education. On average, these children tend to learn better speaking and reading skills when they attend normal schools.

After primary school, most of these children will receive special secondary education. A few will attend a day care centre. Some adolescents with Down syndrome will realise that they cannot keep up with their peers. This can make them insecure, shy and withdrawn. As a result, they may sometimes respond differently than those around them might expect.

Half of all adults with Down syndrome live at home until about the age of thirty. Some people with Down syndrome will live independently in supported accommodation.

They usually live in residential projects for small groups.

The average life expectancy for people with Down syndrome is sixty. People with Down syndrome need guidance and support from their parents and close relatives throughout their lives.

What do their parents and siblings say?

Almost all parents say that they love their Down syndrome son or daughter very much. They are also very proud of their child. Most parents (8 out of 10) feel that their child has given them a more positive outlook on life. The same goes for most siblings. They say that they want to stay involved in their brother or sister's life when they have all grown up. Some families do encounter problems, and often find the situation a strain on the family.

You can read more about this at www.downsyndroom.nl/home/levensloop/kwaliteit-van-leven/.

Health

A Down syndrome baby has a higher than average risk of dying before birth. Almost half of all children with Down syndrome are born with a heart defect. In half of all cases, that will disappear naturally. The other half can almost always be effectively treated by surgery. There are usually no more problems following surgery.

Children with Down syndrome can also suffer from a gastrointestinal abnormality. This can also be treated by surgery.

Children with Down syndrome are at higher risk of respiratory disorders, and of problems with their hearing, eyes, and speech, as well as of impaired immunity to infections. The seriousness of the health problems involved varies from one person to another. Adults with Down syndrome are more likely to develop a particular form of dementia (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 professionals, such as paediatricians, speech therapists, physiotherapists and social workers. Adults with Down syndrome can seek assistance from a physician for people with an intellectual disability, their GP, Down syndrome clinic, or Down syndrome team.

Healthcare insurance will cover the cost of medical care, including any resources – where needed – for children with Down syndrome. There are also various financial aid arrangements for parents.

What is Edwards' syndrome?

Edwards' syndrome is a very serious congenital disorder. It is much less common than Down syndrome. The majority of babies with Edwards' syndrome will die either before or shortly after birth. These babies often show retarded growth before birth. Babies with Edwards' syndrome that are born alive tend to have a low birth weight. Their health is very fragile and they usually die during the first year of life.

Children with Edwards' syndrome suffer from very severe intellectual disability. Approximately 9 out of 10 of these children will have a serious congenital heart defect. In many cases, other organs, such as the kidneys and bowels, are also affected. These children can also suffer from an abdominal wall defect and oesophageal atresia (where the gullet is not connected to the stomach). They may have small faces and large craniums. Their health problems are always serious. The nature (and severity) of these problems can vary from one child to another.

What is Patau's syndrome?

Patau's syndrome is a very serious congenital disorder. It is much less common than Down syndrome. The majority of babies with Patau's syndrome will die either before or shortly after birth. These babies often show retarded growth before birth. Children with Patau's syndrome that are born alive tend to have a low birth weight. Their health is extremely fragile and they usually die during the first year of life.

Children with Patau's syndrome suffer from severe intellectual disability. There are usually serious problems with their brain and heart. Sometimes, they also have kidney disorders and gastrointestinal tract abnormalities. Some of these children may also have extra fingers or toes. They may also have facial abnormalities, such as a cleft lip-jaw-palate (hare-lip). Their health problems are always serious.

The nature (and severity) of these problems can vary from one child to another.

3 It's entirely up to you whether or not you undergo screening

Screening for Down syndrome, Edwards' syndrome and Patau's syndrome is not compulsory. It's entirely up to you. You also decide how far you want to go with the screening test. You can withdraw from the screening test at any time.

Help with choosing

There is a questionnaire at www.onderzoekvanmijnongeborenkind.nl that can help you to organise your own feelings and thoughts on the matter. There are also tips on how to discuss this topic with your partner or others. This discussion with your midwife or gynaecologist will also help you to make a decision.

You may have an increased risk, for instance because you have previously given birth to a baby with Down syndrome, Edwards' syndrome or Patau's syndrome. In that case, you are eligible for a counselling session at a Centre for Prenatal Screening, which is a department in a hospital. There, you will be given detailed information about the available options.

What could the result mean to you?

1. **You will be reassured:** the screening found no sign of any disorders. Or it found a disorder that will not pose a lot of problems in your child's everyday life. Even if the results are good, your baby may still have a disorder, because the screening cannot find all types of disorder.
2. **You become worried:** the result indicates that your baby may be suffering from a disorder. Follow-up diagnostic testing is needed, just to be sure.
3. **There may be difficult choices ahead:** the follow-up diagnostic testing may show that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome. It is also possible that you are carrying a baby with a different abnormality (or chromosomal abnormality). This may force you to make some difficult choices. You must consider what you want to do.

Questions that can help you to reach a decision

- Before your baby is born, do you want to know whether it has Down syndrome, Edwards' syndrome or Patau's syndrome? Or would you rather wait and see?
- How much do you want to know about your baby before it is born?
- Suppose that you get a bad result, and hear that your baby may have a disorder. In that case, would you want to have follow-up diagnostic testing just to make sure? You could decide not to take any further tests and to just carry on with the pregnancy. It's up to you.
- Follow-up diagnostic testing (chorionic villus sampling or amniocentesis) involves a small chance of a miscarriage. How do you feel about these screening tests?
- If the follow-up diagnostic testing shows that your baby does indeed have a disorder, how can you prepare for this?
- How do you feel about life with a child that has Down syndrome, Edwards' syndrome or Patau's syndrome?
- What are your views about the possibility of terminating the pregnancy if the baby has a disorder?
- If you were told that your baby has Down syndrome, Edwards' syndrome or Patau's syndrome, what would you want to do?

4 An in-depth discussion: counselling session

During your first visit to your obstetric care provider, did you indicate that you would like to know more about screening for Down syndrome, Edwards' syndrome and Patau's syndrome?

If so, you will have an in depth counselling session with your obstetric care provider about the screening. You will also be able to ask questions. This is called counselling. The person you will be talking to is called a counsellor.

Feel free to take someone with you

Two people hear more than one. So it's a good idea to bring someone else with you to the counselling session. That could be your partner, for instance, or a female friend, or one of your parents. Avoid bringing any children with you. That way, you will have the chance to talk without interruptions.

After the counselling session, you can decide whether or not you want to participate in the screening.

Your midwife or gynaecologist can help you to reach a decision.

What if, after the counselling session, you are still not sure whether you want to go ahead with the screening? In that case, you can discuss this again with your obstetric care provider. This may help to make things clearer for you. After the in-depth discussion (counselling session), you can decide whether or not you want to go ahead with the screening for Down syndrome, Edwards' syndrome and Patau's syndrome.

5 The screening: a choice of two tests

Have you decided that you want to have the screening for Down syndrome, Edwards' syndrome and Patau's syndrome? If so, then you can choose one of two tests: the NIPT or the combined test.

What is the NIPT?

The NIPT is a screening test in which a blood sample from a pregnant woman is tested. The blood sample is tested in a laboratory. If this screening test shows that the baby may have Down syndrome, Edwards' syndrome or Patau's syndrome, follow-up diagnostic testing is needed, just to be sure. You can have a NIPT from week 11 of your pregnancy.

Scientific study

In the Netherlands, the Non Invasive Prenatal Test (NIPT) is only an option if you are participating in a scientific study (TRIDENT-2). You would have to give researchers permission to use your data. This involves signing a consent form.

At www.meerovernipt.nl you can read more about the scientific study and what happens to your data.

Secondary findings from the NIPT

You can also ask to be informed about any chromosome abnormalities other than Down syndrome, Edwards' syndrome or Patau's syndrome. We call these secondary findings. The laboratory can only find very serious abnormalities in the chromosomes, so any minor abnormalities will not show up. To be clear: the severity of the problem does not just depend on the size of the abnormality. Chromosomal abnormalities usually have serious consequences. If there is a secondary finding, then follow-up diagnostic testing is needed, in order to assess what this means for you or your baby. About 4 in every 1,000 women who opt for NIPT are told that there is a secondary finding.

Take a look at www.onderzoekvanmijnongeborenkind.nl for more information about NIPT and secondary findings. If you opt for NIPT, you should also indicate whether you want to be told about any secondary findings.

What is the combined test?

The combined test is a combination of two screening tests:

1. A blood test given to women in the period from weeks 9 to 14 of their pregnancy. The blood sample is tested in a laboratory.
2. A nuchal translucency test and an ultrasound scan of the baby in the period from weeks 11 to 14 week of the pregnancy. All babies have a thin layer of moisture under the skin in the neck, which is known as the 'nuchal fold'. The thicker the nuchal fold, the bigger the chance that the baby has Down syndrome, Edwards' syndrome or Patau's syndrome.

A list of the differences

Have you decided to screen for Down syndrome, Edwards' syndrome and Patau's syndrome, but you don't know which of the two tests to choose? If so, try using the table on page 15 to make an initial comparison. Studies have shown that the NIPT detects more cases of Down syndrome, Edwards' syndrome and Patau's syndrome than the combined test, and that it is often more accurate. This means that fewer pregnant women are unnecessarily referred for follow-up diagnostic testing.

Your midwife or gynaecologist can give you more information about the tests.

Screening in the case of twins

Are you carrying twins? If so, then you can opt for the combined test and, sometimes, for the NIPT. Discuss this in detail with your midwife or gynaecologist.

Is the pregnant woman's age a factor?

Older pregnant women have a greater chance than younger pregnant women of carrying a baby with Down syndrome. This means that, on average, 19 out of every 10,000 30-year-old pregnant women will be carrying a baby with Down syndrome. On average, 155 out of every 10,000 40-year-old pregnant women will be carrying a baby with Down syndrome. There is also an increased chance that older pregnant women will be carrying a baby with Edwards' syndrome or Patau's syndrome.

A comparison of the combined test and the NIPT

	Combined test	NIPT
What does the test involve?	Blood tests for the pregnant woman, plus a nuchal translucency test and an ultrasound scan for the baby.	The blood test for the pregnant woman.
Does this test involve a scientific study?	No, the test has been used in the Netherlands for quite some time. Y	es, this test has only just been introduced in the Netherlands. You must give permission for your data to be used for this scientific study. See also www.meerovernipt.nl .
When can I take the test?	The blood test can be taken between weeks 9 and 14 of the pregnancy, and the nuchal translucency test between weeks 11 and 14.	From week 11 of your pregnancy.
How long will I have to wait for the results?	This varies from one sonography centre to another. If the blood sample was taken one or two weeks before the ultrasound scan, you will usually be given the result on the day you have the ultrasound scan. If the blood sample was taken later than this, you will usually be given the result several days after you had the ultrasound scan.	Within 10 working days.
Can I choose whether or not to be told about any secondary findings from the test? Y	ou will always be told about any secondary findings.	You can choose whether or not to be told about any secondary findings.
What does the test cost?	€176.96	€175

6 This result does not provide absolute certainty

The results of the NIPT and the combined test do not provide absolute certainty. Nevertheless, in most cases, the result is reassuring: if there is a good result, then there is only a very small chance of an abnormality. In that case, you will not have any follow-up diagnostic testing.

Did you get a bad result? If so, you can opt for follow-up diagnostic testing, just to be sure. Would you like to know more about the reliability of the result? Take a look at www.onderzoekvanmijnongeborenkind.nl.

Who will inform you about the results?

Your obstetric care provider will explain what the result means. What if you decided that you wanted to be told about any secondary findings from the NIPT, and the laboratory has actually reported secondary findings? If so, you will be informed about the results by your obstetric care provider. You may also be called by an expert from a Centre for Prenatal Screening or a university medical centre's clinical genetics outpatient clinic.

When do you get the results?

Exactly when you are told about the result depends on the type of screening test involved. It also differs from one midwife, gynaecologist, and/or hospital to another. You will be informed about this before the screening test in question.

What do the results of the NIPT mean?

If the NIPT result is normal, no follow-up diagnostic testing is needed.

In the event of an abnormal result follow-up diagnostic testing is needed, just to be sure.

What sort of results might you get?

The result is normal.

This result is almost always correct. The chance that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome is very small. Less than 1 in 1,000 pregnant women who get this result turn out to be carrying a baby that has one of these disorders. In that case, no follow-up diagnostic testing is necessary. Your obstetric care provider will inform you of the result.

You have an abnormal result and you may be carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome.

You will be informed about this result by your midwife or gynaecologist. The only way to be sure is to have follow-up diagnostic testing.

- About 90 out of every 100 women who get this result are actually carrying a baby with Down syndrome.
- The corresponding numbers for Edwards' syndrome and Patau's syndrome are 90 and 50 out of every 100 women with abnormal results.

Are you thinking about terminating the pregnancy? In that case, follow-up diagnostic testing is needed first, just to be sure.

A secondary finding has been reported.

Someone will call you and explain what has been found, and what this might mean for you or your baby. You will receive an invitation for a counselling session at a university medical centre's clinical genetics outpatient clinic. During that session, you will be given further information about the secondary finding in question, and about the available options. Follow-up diagnostic testing is needed, just to be sure.

No secondary findings were reported.

The letter with the result states that no secondary findings have been reported. In these cases, no follow-up diagnostic testing is needed, but note: the NIPT cannot find all possible types of chromosome abnormality. For this reason, there is still a slight chance that your baby may, nonetheless, have a disorder.

What do the results of the combined test mean?

The combined test calculates the chance that your baby has Down syndrome, Edwards' syndrome or Patau's syndrome. So this does not provide absolute certainty.

What sort of results might you get?

There is an increased chance that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome.

This means that there is a chance of 1 in 200 (or a bigger chance, e.g. 1 in 50) that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome. You can opt for follow-up diagnostic testing, just to be sure.

There is no increased chance that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome.

This means that the chance you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome is less than 1 in 200 (e.g. 1 in 1000). You will not have any follow-up diagnostic testing. The result of the combined test shows how big the chance is, but it does not give you absolute certainty. So there is a small chance that your baby may, nonetheless, have Down syndrome, Edwards' syndrome or Patau's syndrome.

There are indications that it may have another abnormality or disorder.

If the nuchal fold measures 3.5 millimetres or more, you will always be offered an extensive, additional ultrasound scan. A thickened nuchal translucency is not only associated with Down syndrome, Edwards' syndrome and Patau's syndrome. It can also indicate that a baby has other chromosomal abnormalities and physical disorders, such as heart defects. Sometimes no cause can be found for the thickened nuchal translucency, and the baby is born healthy, with no disorders.

When performing an ultrasound scan, the sonographer sometimes sees other abnormalities in the baby (secondary findings). This could involve serious abnormalities, for instance the limbs or brain might be missing. If you opt for the combined test, you will always be told about any secondary findings.

7 Deciding whether or not to have follow-up diagnostic testing

Is there any evidence that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome? If so, you can always put your questions to your midwife or gynaecologist, or seek their support. If you get a bad result, you could decide not to take any further tests and to just carry on with the pregnancy. But – if you so wish – you can also opt for follow-up diagnostic testing, just to be sure (or to get more clarity). Anyone who is considering terminating their pregnancy at the hospital is always required to have follow-up diagnostic testing first.

It's entirely up to you

If you want more information about follow-up diagnostic testing you can visit a Centre for Prenatal Screening, for a counselling session. After the session, you can decide what you want to do. You may also decide that you don't want to have any follow-up diagnostic testing. The type of follow-up diagnostic testing you will be given depends on several things, such as which type of test you took first.

Was the first test that you took a NIPT?

In the event of a bad NIPT result, the follow-up diagnostic testing may involve either chorionic villus sampling or amniocentesis. With chorionic villus sampling or amniocentesis, you can be sure whether or not your baby has Down syndrome, Edwards' syndrome or Patau's syndrome. The drawback of these screening tests is that there is a small chance they could cause a miscarriage. This happens to 2 out of every 1000 women who take this screening test. However, the other 998 women do not suffer a miscarriage as a result of the test.

- In chorionic villus sampling, a piece of tissue is removed from the placenta and examined. This test can be performed after 11 weeks of pregnancy.
- In the case of amniocentesis, a sample of amniotic fluid is taken and examined. This test can be performed after 15 weeks of pregnancy.

If you would like more information about chorionic villus sampling or amniocentesis, take a look at www.erfaliteit.nl

Was the first test that you took a combined test?

Did the combined test show that there is an increased chance that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome? If so, you can opt for the NIPT or for an invasive screening test (an amniocentesis or chorionic villus sampling). If the NIPT result is not abnormal, you are very probably not carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome. In that case, you will not undergo amniocentesis or chorionic villus sampling. However, if the NIPT result is abnormal, you can still opt for amniocentesis or chorionic villus sampling, just to be sure.

The result of follow-up diagnostic testing

Your specialist will inform you about the results of the follow-up diagnostic testing. The follow-up diagnostic testing may show that there is nothing wrong with your baby. But there is also a chance that these screening tests will show that you are carrying a baby with Down syndrome, Edwards' syndrome or Patau's syndrome. Such news can trigger feelings of sadness or anxiety. You would probably have a lot of questions, too. This means that, soon after the results are known, you will have counselling sessions with one or more medical specialists, such as a gynaecologist, a clinical geneticist, or a paediatrician. Exactly which specialists are involved will depend on the abnormality or disorder that has been found.

Extensive support

You can always count on getting suitable support. A gynaecologist, paediatrician and/or clinical geneticist will talk to you about the results, and about your baby's disorder or abnormality. They will also tell you how this will affect your baby's life and whether it can be treated. There will, of course, be plenty of time for any questions that you and your partner might wish to ask. It is important that you get all the answers you need. You will also be told where to find more information about your baby's disorder or abnormality. Details of the most important organisations and websites are listed on page 24.

Help in deciding what to do about the results

The next step is usually a difficult one – you have to decide what you want to do about the results. It goes without saying that the experts at the Centre for Prenatal Screening will help you with this.

- You can choose to continue the pregnancy. You will then have several months in which to prepare for the arrival of a baby with a disorder or abnormality. You will also have time to tailor the care you receive during pregnancy and delivery, in line with the abnormality or disorder that has been found. Some abnormalities can be treated during pregnancy.
- However, other abnormalities are so severe that the baby may die before or during birth. If this is the case, then the midwife or gynaecologist will give you careful and effective guidance.
- If you find out that your unborn baby has an abnormality, you may find it very difficult to decide what to do. You might consider terminating the pregnancy, for example. Put your questions to your midwife, gynaecologist, paediatrician and/or clinical geneticist. They will help you to reach a decision that is right for you. You will also be offered an opportunity to talk to a social worker, or other experts. If you opt for early termination, this procedure can be carried out up until the 24th week of your pregnancy.

Whatever the result, you should discuss it with your partner, midwife, gynaecologist or GP.

8 Some other things that you need to know

The costs and reimbursements of prenatal screening.

Cost of the in-depth discussion (counselling session)

Your health insurance will cover the cost of the in-depth discussion (counselling session) with your midwife or gynaecologist, about the possibility of screening for Down syndrome, Edwards' syndrome and Patau's syndrome. This will not affect your health insurance's obligatory deductible excess. So you don't have to pay anything yourself.

Cost of the screening

You must pay for the tests yourself. The charge for the combined test is around €177 (in the case of a single baby), while the NIPT costs around €175.

Screening costs where there is an increased chance of a disorder or abnormality

For instance, is there an increased chance that your baby will have Down syndrome, Edwards' syndrome or Patau's syndrome because you have previously given birth to a baby with one of these disorders (see page 10)?

In that case, your care insurer will pay for a counselling session at the Centre for Prenatal Screening and for the test. However, this may affect your obligatory deductible excess.

Ask your care insurer about this.

Costs of follow-up diagnostic testing, and reimbursements

Did you get a bad result in the combined test or the NIPT? If so, you can opt for follow-up diagnostic testing. The costs involved are covered by your basic health insurance package. However, this may affect your obligatory deductible excess. Ask your care insurer about this. The expert from the Centre for Prenatal Screening can also tell you more about this.

Costs and reimbursements can change

The costs and reimbursements described above may change. For up-to-date information on costs, see: www.onderzoekvanmijnongeborenkind.nl/kosten.

Agreement necessary

The counselling session and the prenatal screening will only be available if your care provider has an agreement with a Regional Centre for Prenatal Screening. You should ask about this in advance. You can also check (at www.peridos.nl/zoek-zorgverlener) which obstetric care providers in your region have an agreement of this kind.

Also, ask your care insurer whether your care provider has an agreement with them.

9 More information

Internet

You can find the entire contents of this leaflet, plus some additional information, at www.onderzoekvanmijnongeborenkind.nl. Are you finding it difficult to decide whether you want to have the screening for Down syndrome, Edwards' syndrome and Patau's syndrome? The site's questionnaire may be able to help you with that. There are also tips on how to discuss this topic with your partner or others.

Other websites that provide information on prenatal screening:

www.meerovernipt.nl

www.erfelijkheid.nl

www.deverloskundige.nl

www.thuisarts.nl

www.degynaecoloog.nl

Reading tips

Leaflets containing information about other pregnancy screening tests: there is a separate leaflet containing details of the anomaly scan. That screening test is also part of prenatal screening. You can find that leaflet at www.rivm.nl/20wekenecho/folder. The leaflet entitled 'Pregnant!' contains general information about pregnancy. It also gives details of the blood test that you will take before you are 12 weeks pregnant. These screening tests are used for various purposes, such as identifying your blood group and checking for any infectious diseases. You can find that leaflet at www.rivm.nl/folderzwanger!

You can also ask your midwife, GP or gynaecologist about these 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 decide for themselves whether or not they could handle life with a child with Down syndrome. They will be informed about life with Down syndrome, allowing them to make a decision that is right for them. The Foundation also supports the parents of new-born babies with Down syndrome.
www.downsyndroom.nl

The book 'Downsyndroom – Alle medische problemen op een rij' (Down syndrome - A summary of all the medical problems involved; 2010) summarises the medical problems that commonly affect people with Down syndrome, from the first month of life to the teenage years. The book's authors are Marloes Vegelin (a paediatrician), Paulette Mostart (an editor) and Lex Winkler (a physician and director). It was edited by Michel Weijerman, a paediatrician with extensive experience in the treatment of children with Down syndrome.

Artsen Voor Kinderen (Doctors for Children), Amsterdam, ISBN 987-90-808521-2-9. The book is also available as an app: 'Downsyndroom – Medisch op weg' (Down syndrome – Medical route map), which is available from the app store for iPad and Android.

The Cyberpoli is an interactive internet outpatient clinic for children and young people with a chronic disorder or disability: **www.cyberpoli.nl/downsyndroom**

The Erfocentrum is the national knowledge and information centre for heredity, pregnancy and genetic or congenital defects.

It offers information about Down syndrome, Edwards' syndrome and Patau's syndrome,
www.erfelijkheid.nl, www.zwangerwijzer.nl

VSOP: The Association of Parent and Patient Organisations (VSOP) is involved in genetic issues. It is a partnership of approximately 75 patient organisations, most of which are associated with disorders that are genetic, congenital, or rare. For 40 years, VSOP has been representing these organisations' common interests in the fields of genetic issues, ethics, pregnancy, biomedical research and care for rare disorders. **www.vsop.nl**

Vereniging VG netwerken: connects parents and people with very rare syndromes that are associated with an intellectual disability and/or learning difficulties. www.vgnetwerken.nl

Platform ZON: patient organisation for the parents of children with very rare or unknown disorders, including chromosome abnormalities such as Edwards' syndrome and Patau's syndrome.
www.ziekteonbekend.nl

BOSK: an association for people with limited motor abilities, and their parents. BOSK.nl Dutch Heart Foundation: for more information about congenital heart defects.
www.hartstichting.nl

FetusNed: information about abnormalities in bones, arms or legs and possible treatments:
www.fetusned.nl

RIVM: RIVM coordinates screening programmes for Down syndrome, Edwards' syndrome and Patau's syndrome, and for physical abnormalities, at the request of the Ministry of Health, Welfare

and Sport, in cooperation with various medical professional associations. For more information: www.rivm.nl/down-edwards-patau-seo under 'Organisatie'.

Regional Centres for Prenatal Screening: the eight Regional Centres are all licensed to provide the screening tests described above. They maintain contractual agreements with the screeners and are responsible for regional quality assurance. More information about these Regional Centres can be found at: www.rivm.nl/down-edwards-patau-seo under '**Organisatie**'.

10. How your data is used

If you opt for prenatal screening, the care providers will record data in your healthcare file. Some of this data will also be held in a national database (Peridos). This is necessary to ensure that prenatal screening runs smoothly. The data in Peridos can only be seen by your care providers (such as the midwife, gynaecologist, laboratory worker, nurse and sonographer).

Each region has its own Regional Centre that is licensed by the Ministry of Health, Welfare and Sport (VWS). These Regional Centres ensure that prenatal screening in your region is coordinated effectively. They also monitor all of the care providers involved, to ensure that they perform their duties properly. Staff at the Regional Centres occasionally check the data, to determine whether the screening is going well. The system is highly secure, to protect your privacy.

Scientific research

Having national figures about the use (and effect) of the screening programme is vital, in terms of improving prenatal screening still further. For instance, we keep track of how many pregnant women use prenatal screening and we monitor the results of the various screening tests.

These evaluations and statistics are used by researchers, but the data contains no clues to the identities of the people who took these screening tests.

However, personal data is sometimes necessary for the purposes of scientific research. For instance, when investigating new methods. We would never use your data for that purpose without first asking your permission.

Objection

Would you prefer us not to use your data for evaluation and scientific research? If so, please tell your obstetric care provider. Your data will then be deleted after the expected delivery date. All that will be left is an anonymous report, so that you can be included in the statistics.

Would you like to know more?

Your obstetric care provider can give you more information about the protection of your data. Or go to www.peridos.nl. You can also find more information on these topics at www.onderzoekvanmijnongeborenkind.nl/privacy.

The TRIDENT-1 and TRIDENT-2 studies

Have you opted for the NIPT? If so, then you will be participating in a scientific study. Your data will then be used. Take a look at www.meerovernipt.nl for more information.

Colophon

The contents of this leaflet were developed by a working group. This working group includes representatives from organisations for GPs (NHG), midwives (KNOV), gynaecologists (NVOG), the Regional Centres for Prenatal Screening, sonographers (BEN), paediatricians (NVK), clinical geneticists (VKGN), the Erfocentrum, the Dutch Patient Alliance for Rare and Genetic Diseases (VSOP) and RIVM.
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This document outlines the current situation, based on the most recent available information. The parties responsible for drawing up this leaflet accept no liability for any errors or inaccuracies. For personal advice you should consult your midwife, GP or gynaecologist.

RIVM, May 2019