

Information on screening for Down's syndrome

Prenatal screening

January 2015



Screening for Down's syndrome in brief

- Your obstetrician, GP or gynaecologist will explain the details of the screening programme.
- At the end of this discussion, you decide whether or not you wish to participate in the screening programme.
- Participation in the screening programme is voluntary.
- During the screening programme, the pregnant woman has a blood test, and an ultrasound scan is made of the skin folds in the foetus's neck. Together, these techniques are known as the combined test.
- The result of the combined test indicates the risk of having a child with Down's syndrome (trisomy 21), trisomy 18 and trisomy 13.
- The result of the combined test is merely an indication of the risk, it is not a certainty.
- If you are found to be at increased risk (1 in 200 or above), you can opt for follow-up testing.
- The results can lead to difficult choices, but you can get help with this.

- Children with Down's syndrome (trisomy 21) have an intellectual disability, and often have health problems too. They vary in terms of their developmental potential. Further details can be found in this brochure.
- Children with trisomy 18 and 13 often die around the time of birth, if not before. They rarely live for more than a year. These children have severe physical abnormalities and a severe intellectual disability.

Contents

1. **What information does this brochure contain?** 5
2. **Down's syndrome** 7
 - What is downsyndroom
 - Information on trisomy 18 and trisomy 13
3. **De combined test** 10
 - Blood test and nuchal fold measurement

 - The results indicate the risk
 - How important is the mother's age?
4. **Follow-up testing** 13
5. **Making a conscious decision** 15
 - Help in reaching a decision
6. **Other things that you need to know** 16
 - When can you expect the results?
 - Prenatal screening: costs and insurance coverage
7. **Further details** 18
 - Internet
 - Leaflets and brochures
 - Organisations and addresses
8. **Protection of personal data** 21

Zwanger!

Informatie en adviezen van verloskundigen,
bebaaiers en gynaecologen



Informatie over de 20 wekenecho

Met Inousserief Echoscopisch Onderzoek



1 What information does this brochure contain?

Many expectant parents wonder whether their child will be healthy. Fortunately, the vast majority of children are born healthy. As a pregnant woman living in the Netherlands, you have the option of having your child tested before birth. In this way, you can opt for tests that will tell you how much at risk you are of having a child with Down's syndrome (trisomy 21). You can also obtain details about trisomy 18 (Edwards' syndrome) and trisomy 13 (Patau's syndrome), unless you would prefer not to know.

If you are considering screening for Down's syndrome, you will first have an in-depth consultation with your obstetrician, family doctor or gynaecologist. The information in this brochure can help you prepare for this consultation. You can also reread this information, at your leisure, after the consultation.

The screening may help to set your mind at ease concerning the health of your child. However, it may also disturb you, and confront you with some difficult decisions. You are free to decide whether or not to take these tests, and whether you want follow-up testing in the event of an unfavourable outcome. You can withdraw from the test procedure whenever you wish.

Details of the anomaly scan (the 20-week ultrasound) are given in a separate brochure. This test is also part of the process of prenatal screening. This brochure is available at www.rivm.nl/zowekenecho. You can also ask your obstetrician, family doctor or gynaecologist about this. There is also a leaflet entitled 'Zwanger!' (Pregnant!). This leaflet contains general information about pregnancy, and about blood tests during the 12th week of pregnancy. This includes tests to identify your blood group, and tests for possible infectious diseases.



2 Down's syndrome

What is Down's syndrome?

Down's syndrome (trisomy 21) is a congenital disorder. It is caused by an extra chromosome. Chromosomes, which are present in all of our body cells, contain our genetic characteristics. Each of our cells normally contains two copies of each chromosome. In people with Down's syndrome, each cell has not two but three copies of a specific chromosome (chromosome 21).

Development

Children with Down's syndrome develop more slowly and to a more limited extent than is normal. This varies from one child to another. Nor is it possible to predict how the child will develop. Development should be actively stimulated, and the earlier the better. Young children are usually at home during the day or at daycare. They sometimes go to special day care centres.

Most children with Down's syndrome go to mainstream schools. A small group receives special needs education. The children often make progress in talking, in interacting with others and in learning skills that are important for everyday life. From puberty onwards, increasing numbers of young people with Down's syndrome attend special schools or daycare centres. Adolescents with Down's syndrome are often shy and withdrawn. Compared to other adolescents, they are twice as likely to suffer from attention deficit problems and behavioural problems. The severity of these behavioural problems is directly related to the severity of the intellectual disability involved.

Half of all adults with Down's syndrome live at home until they are about thirty. Others either live in small-scale accommodation or – with support – in a home of their own. Sixty is the average lifespan for people with Down's syndrome. People with Down's syndrome need lifelong support and guidance.

Health

In Down's syndrome, there is a greater than average risk of a miscarriage or of the foetus dying at a late stage of pregnancy. Almost half of all children with Down's syndrome are born with a heart defect. If necessary, this defect can be surgically corrected. The outcome is almost always good.

Children with Down's syndrome may also be born with a gastrointestinal disorder. This, too, requires surgery soon after birth. In addition, children with Down's syndrome are more likely to have problems with their respiratory system, hearing, eyes, speech, and resistance to infections. Adults with Down's syndrome are more likely to develop Alzheimer's disease, and at a younger age, than average.

The severity of their health problems varies from one individual to another.

Children and young people with Down's syndrome, and their parents, can seek assistance from paediatricians, Down's syndrome clinics, or Down's syndrome teams. Down's syndrome teams are composed of various professionals, such as a paediatrician, a speech therapist, a physiotherapist and a social worker. Adults with Down's syndrome can seek assistance from their GP, Down's syndrome clinic, or Down's syndrome team.

Information about trisomy 18 (Edwards' syndrome) and trisomy 13 (Patau's syndrome)

Besides the risk of Down's syndrome, the result of the combined test also provides information about the risk of trisomy 18 (Edwards' syndrome) and trisomy 13 (Patau's syndrome). You will be given this information, unless you have indicated that you do not wish to know the results. The risk of having a child with trisomy 18 and trisomy 13 also increases with the age of the mother. Like Down's syndrome (trisomy 21), trisomy 18 and trisomy 13 are congenital disorders. They are also caused by an extra chromosome. In children with trisomy 18, each cell has not two but three copies of chromosome 18. Children with trisomy 13 have three copies of chromosome 13. Trisomy 18 and trisomy 13 are much less common than Down's syndrome.

Trisomy 18 (Edwards' syndrome)

Children with trisomy 18 have very fragile health. The majority of children with trisomy 18 die during pregnancy, or shortly after birth. Children with trisomy 18 who are born alive usually die during their first year of life.

Children with trisomy 18 have a very severe intellectual disability. About nine out of every ten of these children have a serious congenital heart defect. Other organs, such as the kidneys and intestines, are also often affected. Some may also have an open abdominal wall and a blocked gullet. In trisomy 18 there is often growth retardation, even before birth. As a result, they have a low birth weight. Such children may have small faces and large skulls. While their health problems are always serious, the nature and severity of these problems vary from one child to another.

Trisomie 13 (patausyndroom)

Children with trisomy 13 have very fragile health. The majority of such children die during pregnancy, or shortly after birth. Children with trisomy 13 who are born alive usually die during their first year of life.

Children with trisomy 13 have a severe intellectual disability. There is usually a defect in the development of both the heart and the brain. Sometimes, they also have kidney disorders and gastrointestinal defects. Some may have extra fingers or toes. There is often growth retardation, even before birth. As a result, they have a low birth weight. Such children may also have facial abnormalities, such as a cleft lip-jaw-palate. While their health problems are always serious, the nature and severity of these problems vary from one child to another.

3 The combined test

The *combined* test is used in early pregnancy to find out whether there is an increased chance that your child has Down's syndrome. The test involves no risk for you or your child.

This test consists of a combination of two tests:

1. a *blood test* given to you in the period between weeks 9 and 14 of your pregnancy;
2. a *measurement of the skin fold in your child's neck*. This is conducted by means of an ultrasound scan, in the period between weeks 11 and 14 of your pregnancy.

Blood test and skin fold measurement in the neck

The blood test involves taking a blood sample, which is then analysed in a laboratory. The skin fold measurement in the neck involves an ultrasound scan. In this procedure, the thickness of the skin fold in the neck in your child's neck is measured. This skin fold contains a thin layer of fluid beneath the skin of the neck. This layer of fluid is always present, even in healthy children. The thicker the skin fold in the neck, the greater the likelihood that the child will have Down's syndrome.



The results are merely an indication of the risk

The results of the blood test and the skin fold measurement, combined with your age and the exact duration of the pregnancy, indicate your risk of having a child with Down's syndrome. The test will not provide any certainty.

If you are found to be at an increased risk of having a child with Down's syndrome, you will be offered follow-up testing (see 4).

This follow-up test will clearly show whether your child has Down's syndrome or not.

Increased risk

In the Netherlands, a chance equal to, or greater than one in 200 at the time of testing is regarded as an increased risk. A chance of 1 in 200 means that one in every 200 pregnant women will be carrying a child with Down's syndrome. The other 199 women will not be expecting a child with Down's syndrome. An increased risk is not the same as being at high or great risk.

Even if the test does not indicate that you are at an increased risk, this is no guarantee that your child will be healthy.

Excessive skin fold

An excessive skin fold in the neck is not only associated with Down's syndrome. It is sometimes seen in healthy children too. An excessive skin fold may also indicate other chromosomal and physical disorders in children, such as heart defects. If the test results show a skin fold of 3.5 mm or more, you will automatically be offered an extensive supplementary ultrasound examination.

How important is the mother's age?

The mother's age affects the likelihood of her having a child with Down's syndrome.

The likelihood of having a child with Down's syndrome

The older the mother, the greater the likelihood she will have a child with Down's

Age of woman carrying a child with Down's syndrome	How many of the children with Down's syndrome are detected by the test?
20 – 25	11 to 13 out of 10.000
26 – 30	14 to 19 out of 10.000
31 – 35	20 to 45 out of 10.000
36 – 40	60 to 155 out of 10.000
41 – 45	200 to 615 out of 10.000

Explanation of the table

Nineteen out of every ten thousand pregnant women aged thirty will be carrying a child with Down's syndrome. This means that, in this group, 9981 women will be carrying a child that does not have Down's syndrome. One hundred and fifty-five out of every ten thousand pregnant women aged forty will be carrying a child with Down's syndrome. This means that, in this group, 9845 women will be carrying a child that does not have Down's syndrome.

The combined test in twins

If you are expecting twins, you will receive a separate result for each child. If one or both children have an increased risk of Down's syndrome, you will be offered follow-up testing.

4 Follow-up testing

syndrome. The result of the combined test is merely an indication of the risk. If you are found to be at an increased risk, you can opt for follow-up testing that will provide greater certainty. This follow-up testing consists of chorionic villus testing (between weeks 11 and 14 of your pregnancy) or amniocentesis (after 15 weeks of pregnancy). An extensive ultrasound scan is sometimes carried out. Since 1 April 2014, you also have the option of taking part in a study of the NIPT (Non-Invasive Prenatal Test).

In some cases, you can also opt for chorionic villus sampling or amniocentesis straight away, in a Prenatal Diagnostic Centre. This may be due to a specific medical reason, or because you are aged 36 or above and choose to proceed directly to follow-up testing rather than take the combined test. The details will be discussed in the course of the consultation.

In chorionic villus testing, a piece of placenta tissue is removed and examined. In amniocentesis, a sample of amniotic fluid is taken and tested. Both tests involve a small risk of miscarriage, as a result of the procedures involved. This occurs in three to five of every 1,000 tests carried out. This risk is slightly higher for chorionic villus testing than for amniocentesis.

Would you like further details about chorionic villus testing or amniocentesis? If so, please visit www.erfelijkheid.nl en www.rivm.nl/downscreening.

NIPT study in 2014 and 2015

Since 1 April 2014, you also have the option of taking part in a study of the NIPT (Non-Invasive Prenatal Test). A sample of your blood is taken and is used to examine the DNA of the foetus. In a laboratory, your blood will be tested for Down's syndrome (trisomy 21), trisomy 18 (Edwards' syndrome) and trisomy 13 (Patau's syndrome). The advantage of the NIPT is that no risk of miscarriage is involved. On the other hand, the NIPT does not provide 100% certainty.

The NIPT study will take place during 2014 and 2015. You can only qualify for participation in the NIPT if the results of the combined test show that you are at increased risk (equal to or greater than 1 in 200) of having a child with a trisomy, or if there are medical reasons for doing so. Even if you are aged 36 or above, you will only be eligible for NIPT if the results of the combined test indicate that there is an increased risk of trisomy.

The NIPT results indicate either 'not abnormal' or 'abnormal'.

- An abnormal test result does not necessarily mean that your child has a disorder. If you want more certainty, or if you are considering terminating the pregnancy, further testing is needed to confirm the result of the NIPT.
- In the event of a non-abnormal result follow-up testing is not recommended, as there is only a very small risk that your child actually has a disorder.

Due to the scientific study of NIPT, fewer women than before are being referred for chorionic villus sampling or amniocentesis.

If you require further details, please visit www.meerovernipt.nl.

5 Making a conscious decision

The choice is yours, on whether to proceed with screening for Down's syndrome. If the test reveals an increased risk of having a child with Down's syndrome, trisomy 18, or trisomy 13, you can also decide whether or not you wish to undergo follow-up testing. What should you base your decision on? In this context, you might consider the following topics:

- How much do you want to know about your child before it is born?
- If the combined test were to show that your child may have a disorder, would you want to proceed with follow-up testing?
- How do you feel about the fact that chorionic villus testing or amniocentesis carry an increased risk of miscarriage?
- Follow-up testing may reveal that your child has Down's syndrome. How will you prepare for this outcome?
- How do you feel about life with a child with Down's syndrome (trisomy 21), trisomy 18 (Edwards' syndrome) or trisomy 13 (Patau's syndrome)?
- How do you feel about the possible early termination of a pregnancy involving a child with a disorder?

Follow-up testing may reveal that you are expecting a child with Down's syndrome (trisomy 21), trisomy 18 (Edwards' syndrome) or trisomy 13 (Patau's syndrome). Alternatively, you may be expecting a child with another type of chromosome abnormality. This can present you with some difficult decisions. Discuss this matter with your partner, your obstetrician, family doctor or gynaecologist. If you opt for early termination, this procedure can be carried out up until the 24th week of your pregnancy. If you decide to proceed with your pregnancy, you will be given guidance by your obstetric care worker.

Help in reaching a decision

Do you need help in deciding on whether or not to proceed with screening for Down's syndrome? You can always contact your obstetrician, GP or gynaecologist. Another option is to view the information available at the websites.

6 Other things that you need to know

If you are considering prenatal screening for Down's syndrome, you will first have an in-depth consultation with your obstetrician, family doctor or gynaecologist.

They will then provide you with:

- details about the disorders
- details about the test
- an explanation of the testing procedure
- an explanation of the significance of the results

If you have any questions, make sure to take this opportunity to ask them.

When can you expect the results?

Just how long you will have to wait for the results depends on the nature of the test itself. This also varies from one obstetrician, family doctor and/or hospital to another. You will receive information on when to expect the results before you proceed with the test.

Prenatal screening: costs and insurance coverage

Cost of counseling

Your health insurance will cover the cost of a counselling session with your GP, obstetrician or gynaecologist about the option of screening for Down's syndrome.

Cost of a combined test

You will have to cover the cost of the combined test (see page 10) yourself. If you have supplementary health insurance, this may cover the cost of the combined test. Ask your health insurance company for further details. The cost of the combined test will be covered if you have a medical indication. The cost may have to be covered by your health insurance excess. Ask your health insurance company for further details.

Follow-up testing: costs and insurance coverage

Did the combined test reveal an increased risk of having a child with Down's syndrome (or trisomy 18 or 13) or do you have a medical indication? If so, you will be eligible for follow-up testing (see page 13). The cost of such testing is covered by your basic health insurance package. The cost may have to be covered by your health insurance excess. Ask your health insurance company for further details.

No distinction in terms of age

Until 1 January 2015, pregnant women aged 36 and above had direct access to follow-up testing (chorionic villus sampling or amniocentesis). This no longer applies as of 1 January 2015. Now, like women below the age of 36, they can only choose the combined test. Women aged 36 and above will have to cover the cost of the combined test themselves.

There may be changes in the costs and insurance coverage involved

There may be changes in the costs and insurance coverage described above. For up-to-date information about this see: www.rivm.nl/downscreening > Hoe verloopt de screening > Kosten. You should also always check the conditions of your health insurance policy.

Agreement required

Counselling and combined tests may only be performed by health care providers who have an agreement with a regional prenatal screening centre. The costs of counselling, too, will only be covered if the health care provider has such an agreement. We recommend that you ask your obstetrician, GP or gynaecologist about this in advance. At www.rivm.nl/downscreening > Veel gestelde vragen > Wat kost de combinatietest > Kosten van de screening there is a list of affiliated obstetricians, gynaecologists, or GPs in your region. We also recommend that you check whether your health insurance company has an agreement with the health care provider. Ask your health insurance company for further details.

7 Further details

Internet

The details contained in this brochure can also be found online, at www.rivm.nl/downscreening.

At www.prenetalescreening.nl, you will also find a help centre. In addition, you will find further background information on prenatal screening, follow-up testing, and congenital disorders.

Other websites containing information on prenatal screening:

www.zwangerwijzer.nl

www.nvog.nl

www.knov.nl

www.meerovernipt.nl

Leaflets and brochures

Would you like more details about the tests and disorders described in this brochure?

Ask your obstetrician, GP or gynaecologist for the information fact sheets.

There are fact sheets on:

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

You can also download these information fact sheets at www.rivm.nl/downscreening.

Would you like more details about other tests during and after pregnancy, such as the standard blood test for pregnant women, for blood group and infectious diseases?

Ask your family doctor, obstetrician or gynaecologist for the leaflet entitled "Zwanger!" (Pregnant!) or visit www.rivm.nl/zwanger!

Organisations and addresses

The Erfocentrum

The Erfocentrum is the Dutch information centre on genetics.

www.erfocentrum.nl, www.erfelijkheid.nl, www.zwangerwijzer.nl

Email Erfolijn: erfolijn@erfocentrum.nl

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

Telephone: +31-(0)35-6034040

The Dutch Down's Syndrome Foundation

This is a parents' association that strives to promote the interests of people with Down's syndrome, and those of their parents. The foundation can provide you with further details on Down's syndrome. The foundation also provides support to the parents of newborn children with Down's syndrome.

www.downsyndroom.nl

E-mail: helpdesk@downsyndroom.nl

Telephone: +31-(0)522-281337

Association of VG networks

The Association of VG networks connects individuals with very rare syndromes associated with a mental handicap and/or learning difficulties, and their parents.

www.vgnetwerken.nl

E-mail: info@vgnetwerken.nl

Telephone: +31-(0)30-2727307

National Institute of Public Health and the Environment (RIVM)

The RIVM coordinates screening programmes for Down's syndrome and physical defects at the request of the Ministry of Health, Welfare and Sport, in cooperation with the various medical professional associations. For further details:

www.rivm.nl/downscreening and www.rivm.nl/zowekenecho.

Regional centres

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/downscreening/veelgestelde vragen](http://www.rivm.nl/downscreening/veelgestelde_vragen).

8 Protection of personal data

If you decide to participate in the combined test, 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 are recorded in your own medical dossier, and in the Peridos database. This system is used by all healthcare 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. 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 health care 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.

The same applies to scientific research. Prenatal screening must be continually improved, so scientific research is needed. Scientific research always involves the use of anonymous information. 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.

Would you like more details about how participating in screening affects your privacy? If so, you can read all about it at www.rivm.nl/downscreening under the heading of 'Juridische informatie' (Legal information).

Publishing details

The contents of this brochure were developed by a working group. This working group includes the organisations representing general practitioners (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).

© Centraal orgaan, RIVM

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.

This brochure is also available at www.rivm.nl/downscreening

Design: Vijfkeerblauw - RIVM, January 2015



Rijksinstituut voor Volksgezondheid
en Milieu
Ministerie van Volksgezondheid,
Welzijn en Sport



nvog NEDERLANDSE VERENIGING VOOR
GEBIETENDE EN CYTOMETRIE



RODE KRUIS NEDERLAND

