Information on screening for Down's syndrome

Prenatal screening

2011 version

Contents


Zwanger!
Algemene
informatie
Informatie van verloskundigen, huisartsen en gynaecologen


Informatie over
het Structureel
Echoscopisch
Onderzoek
de 20-weken echo

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. 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. This test can also uncover other disorders. This test is part of a process known as prenatal screening.

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/zwangerschapsscreening. 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). Approximately 180,000 children are born in the Netherlands each year. Each year, about 300 children are born with Down's syndrome.

Mental handicap and health problems

Children with Down's syndrome vary in terms of their developmental potential. All children with Down's syndrome have a mental handicap. This may involve a mild to moderate mental handicap, but it can be severe in some cases. Children with Down's syndrome have a number of specific physical characteristics. Such children develop more slowly than their peers, both physically and mentally. They are also at greater risk of certain physical defects and health problems. The way in which they develop and the severity of their health problems varies from one individual to another.

In the case of children with Down's syndrome, there is a greater than average risk of a miscarriage or of the child dying at a late stage of pregnancy. Almost half of all children with Down's syndrome are born with a heart defect. This defect can usually be treated with surgery, almost always with a good outcome.
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 develop Alzheimer's disease more often, and at a younger age than average.

Recent years have seen major improvements in the care and supervision of people with Down's syndrome. Young children with Down's syndrome and their parents can seek assistance from Down's syndrome teams. These teams are composed of various professionals, such as paediatricians, speech therapists, physiotherapists and social workers. The children and their parents can also use programmes designed to stimulate development.

The parents of children with Down's syndrome often find their own way of dealing with the situation. These days, people with Down's syndrome have a greater chance of enjoying good health than their predecessors. Their life expectancy too has increased. Today, half of those with Down's syndrome can expect to reach the age of sixty.

People with Down's syndrome need lifelong support and guidance.

3 The combined test

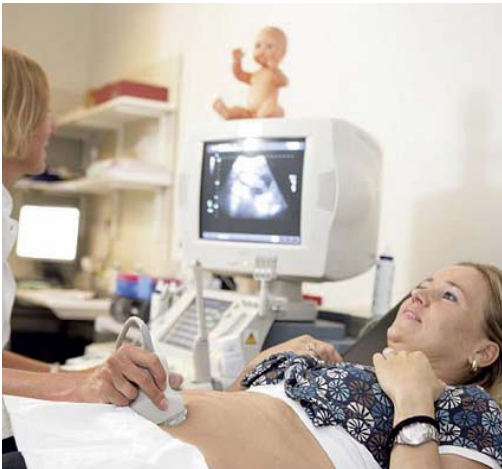
The *combined test* is used in early pregnancy to find out whether there is an increased chance that your child has Down's syndrome. The test involves no risk for you or your child.

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

The blood test involves taking a blood sample, which is then analysed in a laboratory. The skin fold measurement involves an ultrasound scan. In this procedure, the thickness of the skin fold 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, 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 is not only associated with Down's syndrome. An excessive skin fold 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 and the accuracy of the combined test.

The likelihood of having a child with Down's syndrome

The chance of having a child with Down's syndrome increases with the mother's age.

Mother's age	Chance that the mother is carrying a child with Down's syndrome at the time of testing
20 – 25	11 to 13 in 10,000
26 – 30	14 to 19 in 10,000
31 – 35	20 to 45 in 10,000
36 – 40	60 to 155 in 10,000
41 – 45	200 to 615 in 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, 9,981 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, 9,845 women will be carrying a child that does not have Down's syndrome.

The sensitivity of the combined test

The older the mother, the greater the chance of discovering a child with Down's syndrome when using the combined test early in pregnancy. The predictive value of the test is better for older mothers than for young mothers.

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	6 to 7 out of 10
26 – 30	7 out of 10
31 – 35	7 to 8 out of 10
36 – 40	8 to 9 out of 10
41 – 45	9 to 10 out of 10

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.

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

Besides the risk of Down's syndrome, the result of the combination test also provides information about the risk of Patau's syndrome (trisomy 13) and Edwards' syndrome (trisomy 18). 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 Patau's syndrome and Edwards' syndrome also increases with the age of the mother.

Like Down's syndrome, Patau's syndrome and Edwards' syndrome are congenital disorders. They are also caused by an extra chromosome. In children with Patau's syndrome, each cell has not two but three copies of chromosome 13. Children with Edwards' syndrome have three copies of chromosome 18. Patau's syndrome and Edwards' syndrome are much less common than Down's syndrome.

Patau's syndrome

Children with Patau's syndrome have very fragile health. The majority of children with Patau's syndrome die during pregnancy or shortly after birth. Most of these children die during their first year of life.

Children with Patau's syndrome have a serious mental handicap.
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. Health problems are always serious, but the nature and severity of these problems vary from one child to another.

Edwards' syndrome

Children with Edwards' syndrome have very fragile health. The majority of children with Edwards' syndrome die during pregnancy, or shortly after birth. Most of these children die during their first year of life.

Children with Edwards' syndrome have a very serious mental handicap.
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 or oesophageal atresia. In Edwards' syndrome, 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. Health problems are always serious, but the nature and severity of these problems vary from one child to another.

4 Follow-up testing

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. This is known as follow-up testing or prenatal testing.

In some cases, you can also opt for prenatal testing straight away. If you are aged 36 years or over, for example, or if there is a specific medical reason. The details will then be discussed in the course of the consultation.

Chorionic villus testing and amniocentesis

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.prenatalescreening.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, 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, Patau's syndrome or Edwards' 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, Patau's syndrome or Edwards' 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

If you need help in deciding whether or not to undergo Down's syndrome screening, you can discuss this with your obstetrician, family doctor or gynaecologist. Another

option is the internet-based help centre. This helps you to weigh up your options, choices, and concerns. A number of reasons and arguments for and against prenatal screening are given. You can indicate whether or not they apply to you. The help centre then summarises your arguments for and against prenatal screening.
You can find the help centre at **www.kiesbeter.nl/medische-informatie/keuzehulpen/prenatalescreening** and at **www.prenatalescreening.nl.**

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

The in-depth consultation about the test, with your family doctor, obstetrician or gynaecologist is covered by your basic health insurance.

The basic health insurance package will only cover the costs of the combined test if you:

- are 36 or older;
- have another indication for prenatal testing.

If you are younger than 36 and have no other indication, please ask your obstetrician, family doctor or gynaecologist to inform you about the cost of the combined test. If you have supplementary insurance cover, you can ask your health insurance company if this covers the cost of the combined test.

The cost of the consultation and the combined test will only be reimbursed if the individual conducting the screening has an agreement with a regional centre for prenatal screening. We recommend that you ask your obstetrician, family doctor or gynaecologist about this in advance. See www.rivm.nl/zwangerschapsscreening, under “Downscreening” (Down’s screening) and “kosten” (costs), for a list of affiliated obstetricians, gynaecologists or general practitioners in your region. We also recommend that you check whether your health insurance company has a contract with the person conducting the screening. Ask your health insurance company for further details.

Insurance coverage for follow-up testing

If you run an increased risk of having a child with one of the syndromes, you will be eligible for follow-up testing (chorionic villus testing, amniocentesis and/or extensive ultrasound examination). The cost of these tests will then be covered by your health insurance company. In the case of women aged 36 or over, and those with a different indication, the health insurance company will cover the costs of follow-up testing, even if these expectant mothers have not had prenatal screening in advance.

7 Further details

Internet

The details contained in this brochure can also be found online, at **www.rivm.nl/zwangerschapsscreening** and **www.prenatalescreening.nl**. There, 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.zwangernu.nl
www.wijzerzwanger.nl
www.kiesbeter.nl
www.nvog.nl
www.knov.nl

Leaflets and brochures

Would you like more details about the tests and disorders described in this brochure? Ask your obstetrician, family doctor or gynaecologist for the appropriate fact sheets.
There are fact sheets on:

- Second-trimester sonogram (the 20-week ultrasound)
- Down's syndrome
- Patau's syndrome
- Edwards' syndrome
- Spina bifida and anencephaly

You can also download these fact sheets at **www.rivm.nl/zwangerschapsscreening** and **www.prenatalescreening.nl.**

Would you like to know more 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/zwangerschapsscreening.**

Organisations and addresses

The Erfocentrum

The Erfocentrum is the Dutch knowledge and information centre for genetics, pregnancy, and hereditary or congenital disorders.

www.erfocentrum.nl, www.prenatalescreening.nl, www.erfelijkheid.nl, www.zwangernu.nl, www.zwangerwijzer.nl

E-mail 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/zwangerschapsscreening**

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/zwangerschapsscreening/downscreening/kosten.**

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 is 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. If you wish, your personal information can be deleted from Peridos after the screening. Please inform your obstetric care provider about your wishes in this regard.

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. In almost all cases, this involves the use of anonymous information. As many precautions as possible have been taken to ensure that the information cannot be traced back to you or your child. In exceptional cases, scientific research requires data that is traceable. Please tell your care provider if you do not want your data to be used in these exceptional cases.
Whatever your decision, this will not affect the way you are treated before, during or after the screening.

English
This brochure is designed to inform you (and your partner) about prenatal screening for Down's syndrome. The English brochure text is available on ***www.rivm.nl/zwangerschapsscreening***.

Deutsch
Diese Broschüre bietet Ihnen (und Ihrem Partner) Informationen über das pränatale Screening auf Down-Syndrom. Sie finden den deutschen Text der Broschüre auf der Internetseite ***www.rivm.nl/zwangerschapsscreening***.

Français
La brochure vous présente (à vous et à votre partenaire) de plus amples informations sur le dépistage prénatal du syndrome de Down (trisomie 21).
La version numérique de cette brochure est disponible sur le site :
www.rivm.nl/zwangerschapsscreening

Español
Este folleto le ofrece información (también a su pareja) sobre el screening prenatal del síndrome de Down. El texto español de este folleto lo encontrará en
www.rivm.nl/zwangerschapsscreening

Português
Este folheto proporciona a si (e ao seu parceiro) informação sobre o exame pré-natal para detecção da Síndrome de Down. O texto deste folheto em português encontra-se em
www.rivm.nl/zwangerschapsscreening.

Papiamentu
Den e foyeto aki bo (i bo partner) ta haña informashon tokante e screening prenatal di e Síndrome di Down. Bo ta haña e kontenido di e foyeto aki na Papiamentu riba ***www.rivm.nl/zwangerschapsscreening.***

Türkçe
Bu broşür, Down sendromu için doğum öncesinde uygulanan tarama testi hakkında size (ve eşinize) bilgi verme amacıyla hazırlanmıştır. Türkçe metne şu internet sayfasından temin edebilirsiniz: ***www.rivm.nl/zwangerschapsscreening.***

عربي

تم إعداد هذا الكتيّب لتعريفك (وتعريف شريكك) على برنامج العشرين أسبوعاً. يمكن العثور على نص هذا الكتيّب باللغة العربية على الموقع التالي www.rivm.nl/zwangerschapsscreening.

中文
本资料手册致力于为您（和您的伴侣）提供唐氏综合症产前筛查的有关知识。您可以在下面网站上获取本手册的中文版：***www.rivm.nl/zwangerschapsscreening***

Publishing details

The contents of this brochure were developed by a working group. This working group included the organisations representing general practitioners (NHG), obstetricians (KNOV), gynaecologists (NVOG), sonographers (BEN), clinical geneticists (VKGN), the Erfocentrum, the Dutch Genetic Alliance (VSOP) and the 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/zwangerschapsscreening

Obstetricians, gynaecologists, family doctors, sonographers and other obstetric care providers can order additional copies of this brochure through the website www.rivm.nl/pns/folders-bestellen

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

KNOV
KONINKLIJKE NEDERLANDSE
ORGANISATIE VAN VERLOSKUNDIGEN

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nvog
NEDERLANDSE VERENIGING VOOR
OBSTETRIE & GYNAECOLOGIE

VSOP ALLIANTIE VOOR ERFELIJKHEIDSVRAAGSTUKKEN