



Information on the Down's syndrome screening test

Prenatal screening

2009 version

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

Dans cette brochure vous (et votre conjoint ou compagnon) trouverez des informations sur le dépistage prénatal du syndrome de Down. Vous trouverez la version française de cette brochure sur www.rivm.nl/zwangerschapsscreening

Español

Este folleto le ofrece información (también a su pareja) sobre la prueba prenatal sobre el 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 na 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 adresten ulaşabilirsiniz: www.rivm.nl/zwangerschapsscreening.


عربي

تم إعداد هذا الكتيب لتعريفك (وتعريف شريكك) على فحص الوالدين المتعلق بمتلازمة داون. يمكن العثور على نص هذا الكتيب باللغة العربية على الموقع التالي www.rivm.nl/zwangerschapsscreening.

中文

本小手册的·容是要告诉您 (和您的伴侣)
關於·前唐氏症篩檢。您可以在下面網站取得本小手册的中文版，網址：
www.rivm.nl/zwangerschapsscreening.

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1 What can you find in this brochure?



This brochure provides you and your partner with information on prenatal screening for Down's syndrome.

If you are considering prenatal screening for Down's syndrome, you will meet with your midwife, family doctor or obstetrician in advance for an in-depth consultation. The information in this brochure can help you prepare for this consultation. You can also reread all the information at your convenience after the consultation.

What is prenatal screening?

Understandably, many expecting parents wonder whether their child will be healthy. Thankfully, most children are born without any complications. As a pregnant woman in the Netherlands, you are entitled to have your child tested before birth. For example, you can test your likelihood of having a child with Down's syndrome. This test is known as prenatal screening.

Prenatal screening can help set your mind at ease about your child's health. However, it can also be a source of uncertainty and force you to face some difficult decisions. You are free to choose whether or not you want to undergo testing and whether you want follow-up testing if the results are unfavourable. You can decide to break off testing at any time. We also offer a separate brochure on prenatal screening for physical defects, the 20-week ultrasound. This brochure is available at www.rivm.nl/zwangerschapsscreening. You can also ask your midwife, family doctor or obstetrician for a copy.

2 Down's syndrome



What is Down's syndrome?

Down's syndrome is a birth defect. It is caused by an 'extra' chromosome. Chromosomes are in all the cells in our body and contain our genetic characteristics. We normally have two of each chromosome in every cell. People with Down's syndrome have three rather than two of a specific chromosome (chromosome 21) in each of their cells. About two in every 1000 children are born with Down's syndrome. The likelihood of having a child with Down's syndrome increases with the mother's age.

Mental handicap and health problems

People with Down's syndrome are mildly to severely mentally handicapped and have a number of specific physical features. Children with Down's syndrome develop at a slower pace, both physically and mentally. They are also more likely to have certain physical defects and health problems. The child's development and the seriousness of these health problems will differ from person to person.

Some children with Down's syndrome die during childbirth. Almost half the children with Down's syndrome are born with a heart defect. In many cases, this defect can be successfully treated through operation. Some children with Down's syndrome are born with a gastro-intestinal defect, which will also require an operation. They are also more likely to develop health problems related to the respiratory system, hearing, eyes, speech and immunity against infections. People with Down's syndrome are also more likely to develop leukaemia or Alzheimer's disease.

Improved chances of good health

As a result of improved healthcare and better knowledge, people with Down's syndrome now have a greater chance of living a healthy life than they did in the past. Their life expectancy has also improved. In addition, there are now more opportunities for personal development.

3 The combined test

The *combined test* establishes whether your child runs an increased risk of Down's syndrome, during the early stages of pregnancy. The test does not pose any health risks to you or your child.

This test consists of a combination of two tests:

1. *blood test* of the mother, taken between the 9th and 14th week of pregnancy;
2. *measurement of the skin folds in the child's neck*. This measurement is conducted by means of an ultrasound scan between the 11th and 14th week of pregnancy.

Blood test and skin fold measurement

The blood test will involve taking a blood sample, which will then be analysed in a laboratory. The skin fold measurement will involve an ultrasound scan. This scan will measure the thickness of the skin folds in your child's neck. These skin folds contain – both in healthy children and those with Down's syndrome – a layer of fluid. The thicker the skin folds, the greater the likelihood that the child will have Down's syndrome.



The results of the blood test and skin fold measurement, in combination with your age and the exact stage of pregnancy, determine the likelihood of having a baby with Down's syndrome. The test will not provide any certainty. If you have an increased risk of a child with Down's syndrome, you will be offered follow-up testing.

Increased risk

In the Netherlands, a chance of one in 200 or higher is regarded as an increased risk. A chance of 1 in 200 means one out of every 200 pregnant women will be carrying a baby with Down's syndrome at the time of testing. The other 199 women will not be carrying a child with Down's syndrome. An increased risk (one in 200 or higher) is not the same as a high or large risk.

Even if the test results appear to be good, there is still a small chance that your baby will be born with Down's syndrome or another (chromosomal) defect. Testing is no guarantee that you will have a healthy child.

Follow-up testing can determine with certainty whether your child has Down's syndrome or any other chromosomal defect.

Excessive skin folds

In some cases, excessive neck skin folds also occur in healthy children without Down's syndrome. Excessive skin folds may also be a sign of other chromosomal defects and physical disorders. If the test results show a skin fold of 3.5 millimetres or more, you will automatically be offered follow-up testing, even if the results of the combined test do not show any increased risk of Down's syndrome.

What role does the mother's age play?

The mother's age affects the likelihood of 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 average 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 year	1 in 1000
26 – 30 year	1 to 2 in 1000
31 – 35 year	2 to 5 in 1000
36 – 40 year	6 to 15 in 1000
41 – 45 year	20 to 61 in 1000

Explanation of the table

If 1000 30-year-old women are pregnant, 2 of them will be carrying a child with Down's syndrome. This means 998 women will be carrying a child that does not have Down's syndrome.

If 1000 40-year-old women are pregnant, 15 of them will be carrying a child with Down's syndrome. This means 985 women will be carrying a child that does not have Down's syndrome.

Accuracy of the combined test

The average chance that a combined test during the early stages of pregnancy will reveal a child with Down's syndrome increases with the mother's age. The test is less effective in predicting Down's syndrome in young mothers than it is for older mothers.

Age of woman carrying a child with Down's syndrome	How often will the test successfully predict that the woman is carrying a child with Down's syndrome?
20 – 25 year	6 out of 10
26 – 30 year	7 out of 10
31 – 35 year	8 out of 10
36 – 40 year	9 out of 10
41 – 45 year	9 to 10 on 10

Explanation of the table

If 10,000 30-year-old women were pregnant, an average of 20 women would be carrying a child with Down's syndrome. If these 20 were to take the combined test, Down's syndrome would be discovered in 14 of these 20 women, and would not be discovered in 6.

If 10,000 40-year-old women were pregnant, an average of 150 women would be carrying a child with Down's syndrome. If all 150 women were to take the combined test, Down's syndrome would be discovered in 135 of these women, while it would not be discovered in 15 women.

The combined test for multiple births

If you are expecting a multiple birth, you will receive test results for each individual child. If one or more children have an increased risk of Down's syndrome, you will be offered follow-up testing.

4 Follow-up testing

The test results reflect the likelihood, or risk of Down's syndrome. If you run an increased risk, you can decide to undergo follow-up testing in order to be certain. This follow-up testing consists of chorionic villus sampling (between the 11th and 14th weeks of pregnancy) or amniocentesis (after the 15th week of pregnancy). This is known as follow-up testing or prenatal diagnosis, and will help provide more certainty.

In certain cases, you can also choose to undergo a prenatal diagnosis immediately, for example if you are 36 or older, have any genetic or congenital defects in your immediate family or are using medication that could be harmful to your child.

Chorionic villus testing and amniocentesis

Chorionic villus testing involves the removal of a small piece of placenta tissue which is then tested. Amniocentesis involves sampling and testing of the amniotic fluid.

Both tests involve a small risk of miscarriage. This occurs in three to five of every 1000 women to receive testing. The risk level for chorionic villus testing is slightly higher than that for amniocentesis.

If you would like more information on chorionic villus testing or amniocentesis, please visit www.prenatalescreening.nl

5 Making a conscious decision

You are free to decide whether or not you want to undergo Down's syndrome screening. 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 I base my decision on?

- How much do you want to know about your child before it is born?
- If the combined test reveals that your child may have Down's syndrome, would you have follow-up testing carried out?
- Follow-up testing may reveal that your child has Down's syndrome. How will you prepare for this outcome?
- Can you imagine what life would be like if your child had Down's syndrome, and how do you think you would deal with this?
- How do you feel about ending the pregnancy prematurely if your child has Down's syndrome?

Follow-up testing may reveal that you are carrying a child with Down's syndrome. You may then be forced to make some difficult choices. Discuss this with your partner and your midwife, family doctor or obstetrician. If you do decide to terminate the pregnancy prematurely, you may do so until the 24th week of pregnancy.

Help in making a decision

If you need help in deciding whether or not to undergo Down's syndrome screening, you can discuss this with your midwife, family doctor or obstetrician. You can also consult the special Internet help centre. This help centre allows you to weigh your options, choices and objections against one another so that you can make a balanced decision. The help centre will list a number of arguments for and against prenatal screening, so that you can select whether or not they apply to you. The help centre will then provide you with a clear overview of your arguments for and against prenatal screening. The help centre is available at www.prenatalescreening.nl and www.kiesbeter.nl/medischeinformatie/keuzehulpen

6 What else do I need to know?

If you are considering prenatal Down's syndrome screening, you will meet with your midwife, family doctor or obstetrician in advance for an in-depth consultation. They will then provide you with:

- information on the test
- an explanation of the testing procedure
- an explanation of the various possible results
- information on various issues, including Down's syndrome

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

When can I expect the results?

This depends on which test you will be undergoing and may differ depending on your midwife, family doctor and/or hospital. You will receive information on when to expect the results before testing.

Prenatal screening: costs and insurance coverage

This in-depth consultation with your family doctor, midwife or obstetrician is covered by your basic health care insurance.

The combined test is only covered by your basic health insurance if:

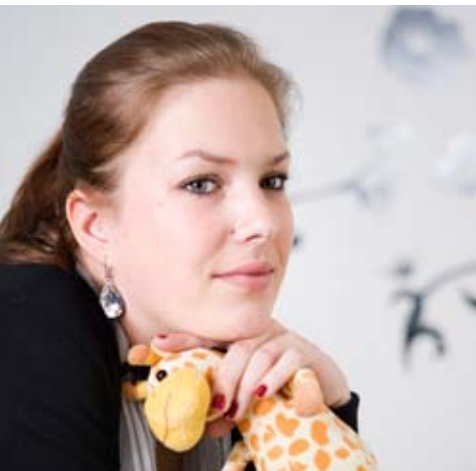
- you are 36 or older
- you have an official doctor's recommendation for prenatal diagnosis

If you are younger than 36 and do not have an official doctor's recommendation, your midwife, family doctor or obstetrician will be able to provide you with more information on the costs of the combined test.

The costs of the consultation and combined test will only be covered by your health insurer if the person carrying out the test is affiliated with a regional centre for prenatal screening. We recommend that you ask your midwife, family doctor or obstetrician about this in advance. You can visit www.rivm.nl/zwangerschapsscreening and click on 'Downscreening' and 'kosten' (costs) for an overview of the affiliated midwives, family doctors and obstetricians in your region. We also recommend that you check whether your health insurer has a contract with the person conducting the screening. Ask your health insurer for more information.

Insurance coverage for follow-up testing

If you run an increased risk of having a child with Down's syndrome, you will be eligible for follow-up testing (chorionic villus testing or amniocentesis). The costs of these tests are covered by your health insurer. If you are 36 or older and/or have an official doctor's recommendation, your health insurer will cover the costs of follow-up testing even if you have not had prenatal screening in advance.



7 Further information

Internet

The information in this brochure is also available online, at www.rivm.nl/zwangerschapsscreening and www.prenatalescreening.nl. These sites also feature a help centre to help you make a balanced decision. You will also find more background information on prenatal screening, follow-up testing and birth defects. Other websites featuring information on prenatal screening:

www.zwangernu.nl

www.kiesbeter.nl

www.nvog.nl

www.knov.nl

Leaflets and brochures

If you would like to know more about the tests and birth defects discussed in this brochure, ask your midwife, family doctor or obstetrician for information leaflets.

Information leaflets are available on the following subjects:

- The combined test
- 20-week ultrasound
- The triple test
- Down's syndrome
- Spina bifida and anencephaly

You can also download these information leaflets at

www.rivm.nl/zwangerschapsscreening and www.prenatalescreening.nl

You may want more information on other tests during and after pregnancy, such as the standard blood test for pregnant women to determine blood type and detect infectious diseases. If so, ask your midwife, family doctor or obstetrician for the Pregnant! brochure, or visit www.rivm.nl/zwangerschapsscreening

Organisations and addresses

The Erfo-centre

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

www.erfocentrum.nl, www.prenatalescreening.nl, www.erfelijkheid.nl,

www.zwangernu.nl, www.zwangerwijzer.nl

Erfo helpline e-mail address: erfolijn@erfocentrum.nl

Erfo helpline: 0900 – 66 555 66. The Erfo helpline is open on Mondays and Thursdays, from 10.00 to 15.00 (€ 0.25 a minute).

Stichting Downsyndroom

Stichting Downsyndroom (Down's Syndrome Foundation) is a parents' association that strives to promote the interests of people with Down's syndrome and their parents. The foundation can provide you with further information on Down's syndrome. The foundation also provides support to parents of new-born children with Down's syndrome.

[www.Downs syndrome.nl](http://www.Downsyndrome.nl)

E-mail: helpdesk@Downsyndrome.nl Phone number: 0522 - 28 13 37.

RIVM

The RIVM (Dutch National Institute for Public Health and the Environment) 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 more information, visit:

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.nl/pns/down-seo/organisatie

8 Protection of personal data

This chapter describes the procedures for protecting the personal data gathered during prenatal screening.

If you decide to have your unborn child tested, its personal data will be stored in a medical dossier. This personal data includes the image scans created during screening. By granting permission for testing, you are also granting permission for the collection and use of this data. Without this data, it would not be possible to make an accurate diagnosis or offer effective treatment. The Medical Treatment Contracts Act obliges care providers to handle this data with the utmost care. For example, the Act specifies that personal data may only be accessed by those involved in the treatment of the person concerned.

Compliance with the applicable quality standards is an essential part of the screening procedure. In order to allow for the assessment of this quality, data from the medical dossier may be made available to the parties responsible for monitoring quality, unless you object to your midwife or obstetrician. If you do object, this will not affect the way in which you are treated before, during or after the screening in any way.

In addition to the monitoring of screening quality, there are also strict legal requirements with regard to the quality of the midwife, family doctor or obstetrician and the regional centre conducting the screening programme. This will be enforced by means of regular quality checks. Scientific research will also be conducted on a regular basis in order to ensure that the programme still meets the latest requirements and to develop new methods of treatment. This research will also require your personal data; however, your data will only be used if you have given permission. If you have granted permission for testing, the midwife, family doctor or obstetrician will ask whether you also agree to your data being used for these purposes.

If you do object, this will not affect the way in which you are treated before, during or after the screening in any way. However, permission to use this personal data for the purposes described above would be highly appreciated, as the test results are of importance to all other pregnant women as well as yourself.

Colophon

The contents of this brochure were developed by a working group. This working group includes the association of family doctors (NHG), midwives (KNOV), obstetricians (NVOG), ultrasound operators (BEN), the Erfo-centre, the Dutch Association of Parent and Patient Organisations (VSOP) and the RIVM.

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

This brochure on Down's syndrome screening is also available at www.rivm.nl/zwangerschapsscreening

Midwives, obstetricians, family doctors, ultrasound operators and other obstetrical care providers can order additional copies of this brochure through the website www.rivm.nl/pns/folders-bestellen

Layout and design: Uitgeverij RIVM, December 2008

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KNOV
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ORGANISATIE VAN VERLOSKUNDIGEN


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B beroepsvereniging
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