

Contents

Why has prenatal testing been offered to me?	2
What fetal abnormalities are tested for in pregnancy?	3
Chromosomal abnormalities	3
About Down syndrome	4
About Edwards syndrome	4
About Patau syndrome	4
About sex chromosomal abnormalities	4
Physical abnormalities	5
What are the different types of prenatal testing?	6
Screening tests	6
Understanding screening results when they are in words	9
Understanding screening results when they are numbers	10
What happens after the screening test?	11
What if I get an “increased risk” result on any screening test?	11
Support available after screening tests	11
Diagnostic testing	12
Chorionic Villus Sampling (CVS)	12
Amniocentesis	13
What happens after a diagnostic test?	14
What if an abnormality is diagnosed from testing?	14
Women’s stories	15
Resources	16
Prenatal Diagnosis and Genetic Counselling Services	16
Web sites	16
Work Sheet	17
Summary of Tests	Inside back cover



Introduction

The aim of this Decision Aid is to assist women in early pregnancy to make decisions regarding testing for fetal abnormalities like Down syndrome.

This Decision Aid will be useful to you if:

- you are interested in learning more about prenatal testing
- you are undecided whether or not to have testing or
- you are unsure about which test to have.

The Decision Aid is designed to be flexible to meet your individual information needs.

- The table inside the back cover gives you a brief summary of the tests available.
- The booklet provides information on the conditions that can be tested, the benefits and risks of the tests, how to make sense of the results and the resources available.
- The worksheet at the back of this booklet is designed to help you weigh up the options.

Why has prenatal testing been offered to me?

- Prenatal testing is made available because of the small risk that all women have of having a baby with a major problem.
- While the majority of babies are born without any abnormality, 3–4% of pregnancies result in babies with abnormalities.

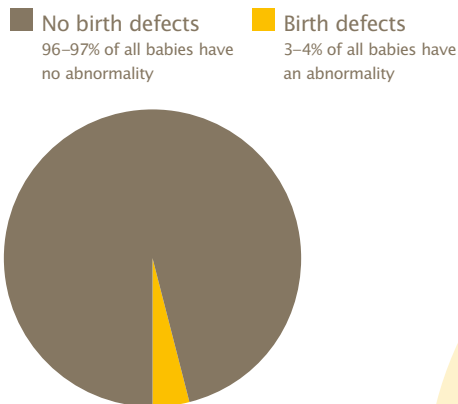


Figure 1: Background risk of birth defects

- Information on prenatal testing for abnormalities in the developing fetus is provided to all women in pregnancy.

- Some but not all fetal abnormalities can be detected during pregnancy.
- No single test checks for everything.
- There are different types of tests available. They vary in how accurate they are, how safe they are, when they are performed and what information they give.
- A woman may decide to have testing:
 - because she wants to know more about her pregnancy,
 - because she wants the option of terminating an affected pregnancy or
 - so she can prepare herself for a baby with an abnormality.
- It is important to understand that ultrasound is a test for fetal abnormality.
- A woman may decide not to have any prenatal testing.

Testing for fetal abnormality is not compulsory for anyone.

What fetal abnormalities are tested for in pregnancy?

- It is not possible to test for all fetal abnormalities in pregnancy.
- It is important to remember that the vast majority of babies born, regardless of a mother's age, do not have any of these abnormalities.

Chromosomal abnormalities

- For normal human development we need 46 chromosomes (23 pairs).
- Any extra or missing chromosome, or any change in the structure or arrangement of the chromosomes in the developing embryo, may affect normal development.
- The chromosomal abnormalities that are most commonly tested for in pregnancy result from an extra or missing chromosome in the developing fetus.
- The risk of these conditions caused by an extra chromosome (eg Down syndrome) increases with a woman's age (Figure 2), but pregnancies of younger women can also be affected.

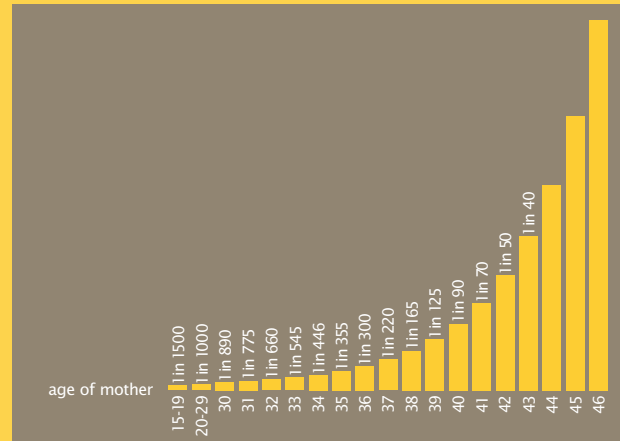
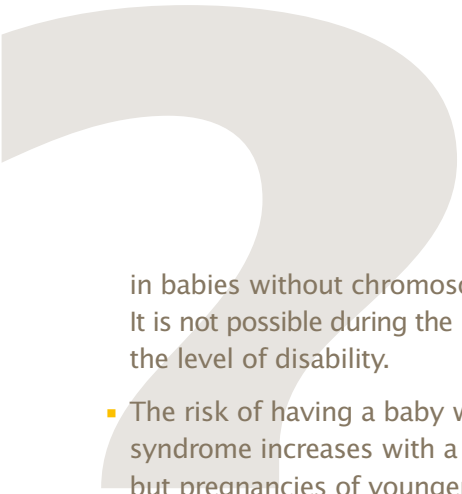


Figure 2: Risk of having a baby born at term with Down syndrome by age of mother

About Down syndrome (also called Trisomy 21)

- Down syndrome is the most common chromosomal abnormality (1 in every 400 pregnancies). It is caused by having an extra chromosome (chromosome 21) and results in a number of characteristic physical, medical and intellectual features.
- Many pregnancies with this chromosomal abnormality miscarry.
- Most individuals with Down syndrome have a mild to moderate level of intelligence and can participate in school, work and social life but others can be severely intellectually disabled. Physical abnormalities involving the heart and digestive system occur more often with babies with Down syndrome than



in babies without chromosomal abnormalities. It is not possible during the pregnancy to predict the level of disability.

- The risk of having a baby with Down syndrome increases with a woman's age but pregnancies of younger women can also be affected.
- There is no cure for Down syndrome but with early intervention such as individualised educational programs, the chances of the child achieving his or her potential is maximised.

For further information go to the Down syndrome Association of Victoria website: <http://www.dsav.asn.au/>

About Edward syndrome (also called Trisomy 18)

- Edwards syndrome is less common than Down syndrome (1 in every 1600 pregnancies) and is caused by having an extra chromosome (chromosome 18).
- Pregnancies with this chromosomal abnormality usually miscarry and babies that are liveborn rarely survive for long.
- All babies with Edwards syndrome have significant developmental problems.
- The risk of having a pregnancy affected with Edwards syndrome increases with a woman's age but pregnancies of younger women can also be affected.

About Patau syndrome (also called Trisomy 13)

- Patau syndrome is less common than Edwards syndrome (1 in every 3400 pregnancies) and is caused by having an extra chromosome (chromosome 13).
- Pregnancies with this chromosomal abnormality usually miscarry and babies that are liveborn are not expected to live.
- All babies with Patau syndrome have severe developmental problems.
- The risk of having a pregnancy affected with Patau syndrome increases with a woman's age but pregnancies of younger women can also be affected.

About sex chromosomal abnormalities

- The chromosome count for typical development of a girl is 46XX and for a boy is 46XY.
- Sex chromosomal abnormalities occur when there is a missing or an extra copy of the X chromosome or an extra copy of the Y chromosome.
- Normal development is affected in a variety of ways depending on the chromosomal abnormality.
- These abnormalities are not known to be associated with a woman's age.

Regardless of a woman's age, the risk of having a pregnancy with a chromosomal abnormality decreases as the pregnancy develops. This is because many pregnancies affected with chromosomal abnormalities will miscarry naturally during pregnancy, without any intervention.

- It is not possible to accurately predict which pregnancies will miscarry.

Physical abnormalities

- Some, but not all, physical abnormalities can be detected during pregnancy.
- Many of the abnormalities that are detected can be treated once the child is born.
- Most physical abnormalities are not related to the mother's age, but some can be related to chromosomal abnormalities or other syndromes.

The most common physical abnormalities include:

- Neural tube defects such as spina bifida. These occur early in pregnancy when the spine fails to form properly and the spinal cord and/or brain can be affected. Taking folate before and during early pregnancy is known to reduce the occurrence of neural tube defects.

For further information go to the websites

of the Public Health Association of Australia and the Spina bifida Foundation of Victoria: <http://www.phaa.net.au/policy/folate> and <http://www.sbfv.org.au>

- Cardiac abnormalities such as conditions involving the development of the blood vessels or the heart's structure.
- Digestive system abnormalities where there is a narrowing or missing parts of the system.
- Renal abnormalities such as blockages or poor development of the kidneys.

It is important to remember that the vast majority of babies born, regardless of a mother's age, do not have any of these abnormalities.

What are the different types of prenatal tests ?

The tests available to check for abnormalities in pregnancy fall into two groups: screening tests and diagnostic tests.

- **Screening tests** can be blood tests from the pregnant woman and/or ultrasounds of the fetus. These tests are available to women of all ages.
- **Diagnostic tests** are tests that examine samples of the developing placenta or fluid taken from around the baby. A skilled obstetrician should do this by inserting a needle into the woman's abdomen to take the sample, guided by ultrasound. These tests are available to women who have been identified as being at "increased risk" because of their age or the results of screening tests.

Screening tests

- All pregnant women should be given appropriate written information and offered prenatal screening, regardless of their age.
- Screening tests do not have any additional risk of miscarriage.
- All screening tests give a risk result not a diagnosis. The result can be in words or numbers or both.
- Screening tests will miss some pregnancies with Down syndrome. Detection rates vary depending on the test and the age of the woman.
- Screening tests are more accurate in women who are older.

Combined First Trimester Screening

The information from two screening tests are combined with the woman's age and gestation to provide a risk estimate for Down syndrome and Edwards syndrome. These tests performed early in pregnancy are:

1. blood taken from the pregnant woman between 8 to 12 weeks that measures two hormones in the woman's blood
2. an ultrasound scan (nuchal translucency) from 11 and a half weeks to the end of the 13th week that measures the thickness behind the baby's neck.

- Combined First Trimester Screening identifies more pregnancies with Down syndrome than using nuchal translucency alone.
- Figure 3 shows how accurate the Combined First Trimester Screening is in detecting Down syndrome. 85–90% of pregnancies with Down syndrome are detected and about 10% – 15% are missed.

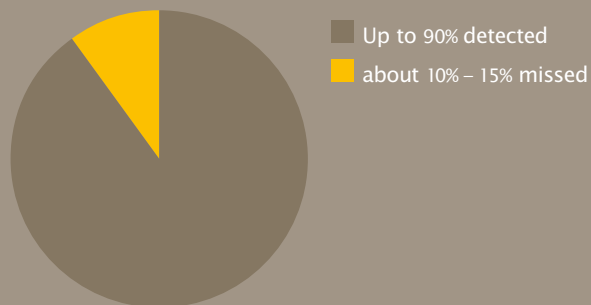


Figure 3: Detection rate of Down syndrome using Combined First Trimester Screening

- These detection rates are based on the ultrasound being done by a person who has had extra training and is recognised as practising at a high standard. For a list of registered centres: www.nuchaltrans.edu.au
- Ultrasound uses sound waves to produce an image (see Figure 4).

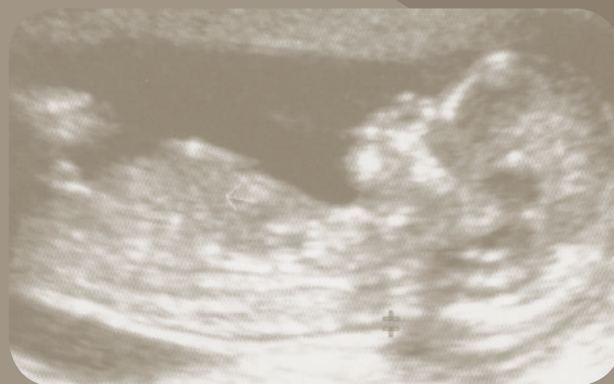


Figure 4: Image of a Nuchal Translucency scan

- This early ultrasound can also:
 - confirm the due date
 - identify twins
 - identify if a miscarriage has occurred.
- The fetus is between 4.5 and 8.4 cms long at this early stage of pregnancy. The ultrasound can only detect some major abnormalities as the physical features of the fetus cannot be seen well. This ultrasound does not replace the detail available from a later ultrasound.
- It is important to keep in mind that most pregnancies identified as at “increased risk” for Down syndrome will not have Down syndrome.

Screening test costs

Screening test	Medicare rebate	Out of pocket costs
Nuchal Translucency alone	\$88.70	\$60-100
Combined First Trimester Screening	\$122.10	\$100-150

Note 1: These costs are estimates only.

Note 2: Medicare rebate relates to ultrasound requested to screen for fetal abnormality.

Second Trimester Maternal Serum Screening

Blood is taken from the pregnant woman between 14–20 weeks (best between 15–17 weeks).

- The test measures the levels of four hormones in the woman's blood and is combined with the woman's age, weight and weeks of pregnancy. It is important that pregnancy dates are accurate.
- Maternal Serum Screening gives a risk estimate for chromosomal abnormalities (Down syndrome, Edwards syndrome) and neural tube defects. The test results can also indicate that a pregnancy requires extra assessment such as another ultrasound.
- The Maternal Serum Screening test can detect up to 75–80% of pregnancies with Down syndrome, missing 20–25% (see Figure 5 below).

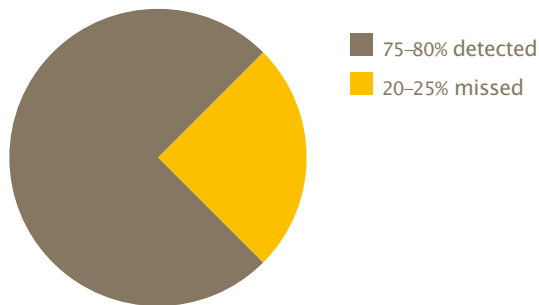


Figure 5: Detection rate for Down syndrome using Second Trimester Maternal Serum Screening

Screening test costs

Screening test	Medicare rebate	Out of pocket costs
Maternal Serum Screening	\$46.35	\$30–80

Note 1: A woman attending a public hospital as a public patient is not charged

Note 2: These costs are estimates only.

Second Trimester Ultrasound

An ultrasound examination between 18–20 weeks of pregnancy.

- Many fetal physical abnormalities can be detected on this later ultrasound. Where abnormalities can be treated, early detection may improve outcomes for the baby.
- Risk factors for chromosomal abnormalities can also be detected on this ultrasound. Sometimes called 'soft markers', these are findings on the ultrasound that do not give definite information but may indicate the increased risk of a problem. A diagnostic test may be required to exclude a chromosomal abnormality.
- This ultrasound is not a reliable test for Down syndrome.
- Other information from this ultrasound includes:
 - The growth and well being of the fetus
 - The position of the placenta
- Not all physical abnormalities can be identified.

Screening test costs

Screening test	Medicare rebate	Out of pocket costs
Second Trimester Ultrasound	\$85	\$80-100

Note 1: Women attending some public hospitals as public patients may not be charged

Note 2: These costs are estimates only

Understanding screening results when they are in words

- The results of screening tests **do not** give “no” or “yes” answers. The risk results use words like “no increased risk” or “increased risk,” and/or numbers like 1 in 1000 or 1 in 200.
- Risks are based on a cut off level to determine whether or not further investigation with a diagnostic test is indicated.
- The risk cut off level for Down syndrome is 1 in 250, or 1 in 300 depending on the test.
- If a test result is “low” or “not increased” risk, it does not mean no risk, but it does mean that diagnostic testing is not indicated.

- In order for screening tests to have the highest detection rate possible, the laboratory sets a cut-off level that makes about 5% (1 in 20) of all pregnancies tested fall into the “increased risk” category for Down syndrome. The majority of these results will be in pregnancies that are not affected with Down syndrome (Figure 6), but a diagnostic test would be needed to find this out in pregnancy.

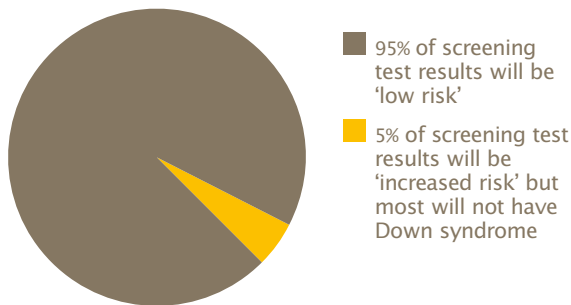


Figure 6: Percentage of screening test results that are “increased risk”

- Of the pregnancies identified as at “increased risk” for Down syndrome only 2% will be affected.

Understanding screening results when they are numbers

- For all risk results it is important to understand what the numbers are telling us about the chance of a fetal abnormality as well as the chance of not having a fetal abnormality.
- Some women find it easier to understand numbers visually as diagrams. If we think of a risk result for Down syndrome such as 1 in 100, this also means a chance of 99 out of 100 the pregnancy will **not** be affected. This may be easier to understand by looking at the diagrams below.

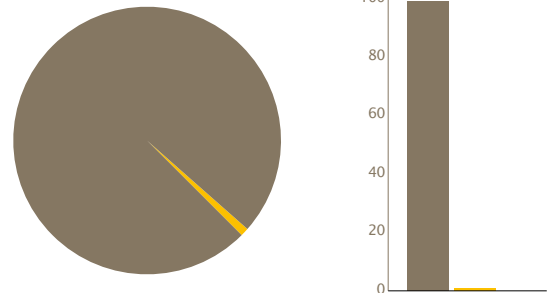
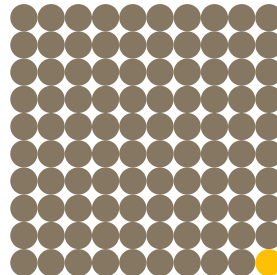


Figure 7: A risk of 1 in 100 for Down syndrome means that out of 100 pregnancies, 1 has Down syndrome and **99 of the 100 do not.**



What happens after the screening test?

- Many women find waiting for results an anxious time. It may be useful to ask the doctor ordering your blood test and/or ultrasound:
 - when you should expect to get the result?
 - who will let you know the result?
 - how will you be notified (ie by letter, phone call or at your next visit)?
- The waiting time for screening tests usually takes up to one week.
- If the results of the screening test estimate “no increased risk for Down syndrome” then no further testing is indicated.

What if I get an “increased risk” result on any screening test?

- If an “increased risk” result is received, it is important to remember that it is more likely that the fetus **will not** have a chromosomal abnormality.
- A diagnostic test is offered to provide reassurance that the fetus does not have a chromosomal abnormality or to confirm that the fetus is affected.

- An “increased risk” result from a Combined First Trimester Screening means the early diagnostic test (CVS) is available (see page 12).
- An “increased risk” result from Second Trimester Maternal Serum Screening means the later diagnostic test (amniocentesis) is available (see page 13).

Support available after screening tests

- If you need a more detailed explanation of your screening test result, it is advisable to make another appointment with the doctor who ordered the test or the health professional providing your pregnancy care. Other options available to you include contacting the genetic counsellors at one of the organisations detailed on page 16.

Diagnostic tests

- There are two diagnostic procedures available: chorionic villus sampling (CVS) and amniocentesis. Both procedures confirm whether there is or isn't a chromosomal abnormality.
- These procedures have different miscarriage risks and are performed at different stages of pregnancy.

Chorionic Villus Sampling (CVS)

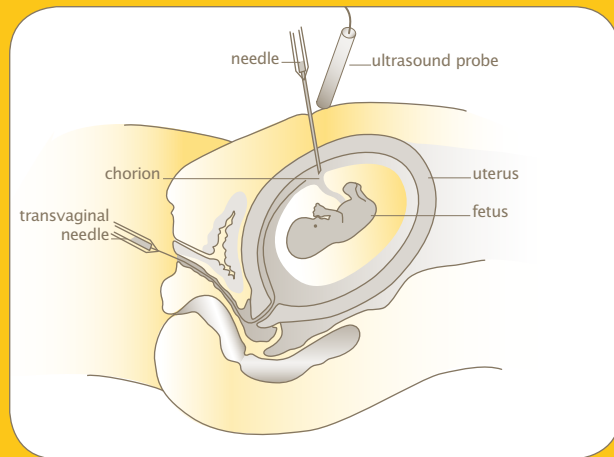


Figure 8: Cross section of abdomen showing chorionic villus sampling (CVS)

- This procedure involves the insertion of a fine needle through the woman's abdomen. Under the guidance of an ultrasound, cells from the developing placenta are obtained and sent to the laboratory to be grown and analysed.
- A CVS is only performed through the vagina if the position of the placenta prevents an abdominal approach.
- CVS is the earlier diagnostic procedure, available from 10–13 weeks of pregnancy and can give a definite diagnosis of chromosomal abnormalities.
- CVS adds a further 1% risk of miscarriage to the risk of miscarriage for all pregnancies.
- On very rare occasions, because of difficulties with growing the cells or with analysing the results, another test may be required.
- CVS is available to women identified as being at an “increased risk” of a chromosomal abnormality like Down syndrome, on the basis of screening test results or their age.
- If a chromosomal abnormality is confirmed, a woman may choose to continue with the pregnancy or to terminate the pregnancy. Support is available for a woman making either decision.
- The option of termination of the pregnancy is a surgical procedure at this stage of pregnancy, without having an induced labour.

Amniocentesis

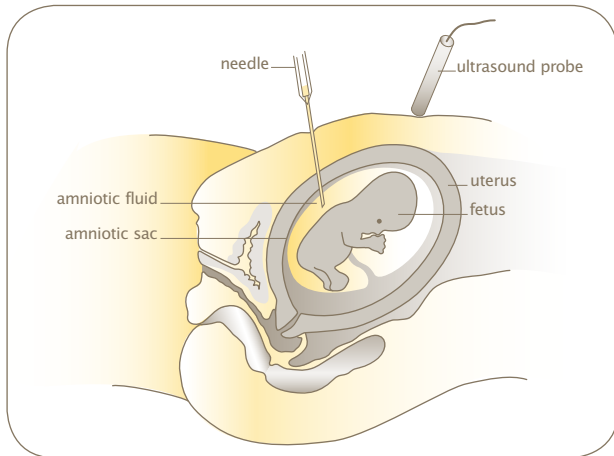


Figure 9: Cross section of abdomen showing amniocentesis

- Amniocentesis involves the insertion of a fine needle through the woman's abdomen. Under the guidance of an ultrasound, a small sample of the fluid from around the fetus is obtained and sent to the laboratory for the fetal cells to be grown and analysed.
- This is the later diagnostic procedure and is done from 15–19 weeks. It can give a definite diagnosis of chromosomal abnormalities.
- The miscarriage risk related to an amniocentesis is less than the CVS: an additional 0.5% miscarriage risk to the risk of miscarriage that all pregnancies have.

- On very rare occasions, because of difficulties with growing the cells, another amniocentesis may be required.
- Amniocentesis is available to women identified as being at an “increased risk” of a chromosomal abnormality like Down syndrome, on the basis of screening test results or their age.
- If a chromosomal abnormality is confirmed, a woman may choose to continue with the pregnancy or to terminate the pregnancy. Support is available for a woman making either decision.
- The option of termination of the pregnancy is most likely to involve labour being induced at this stage of pregnancy.

Diagnostic test costs

Diagnostic test*	Medicare rebate	Out of pocket costs
CVS	\$502.10	\$224 - \$260
Amniocentesis	\$460.10	\$224 - \$270

*(incl laboratory costs)

Note: Women identified as “at increased risk” attending tertiary public hospitals as public patients may not be charged.

What happens after a diagnostic test?

- The final results of a CVS or an amniocentesis can take up to two weeks. In some cases a preliminary short-term result can be made available in 24–48 hours (FISH test).
- Waiting for these results can be an anxious time so it is advisable to ask the doctor doing the test:
 - when you should expect to get the results?
 - who will let you know the results?
 - how will you be notified (ie by letter or phone call?)
- If no abnormalities are detected then no further testing for chromosomal abnormalities is required.

What if an abnormality is diagnosed?

- A woman's choice to continue with the pregnancy or to have a termination of pregnancy will be respected and supported.
- If the woman chooses to continue with the pregnancy, referral to one of the services on page 16 will provide specialised information and counselling. This may include arranging contact with a support group as needed. For details of support groups go to the Genetic Support Network Victoria website:
<http://www.gsnv.org.au>

- If the woman chooses to terminate the pregnancy, the method used will depend on the woman's stage of pregnancy.
- Early in pregnancy the procedures used are called a Dilatation and Curettage (D & C) or a Dilatation and Evacuation (D & E). The procedure is performed under anaesthetic and the woman usually goes home the same day.
- Later in pregnancy the procedure used involves labour being induced. It may take 2–3 days. Different types of pain relief are used according to individual needs.
- Following the termination, follow up care is important to assist in the woman's physical and emotional recovery.

Women's stories

The stories of four women below, describe women's choices regarding prenatal testing for fetal abnormalities. The women have all made different decisions based on their views on accuracy, safety and timing of the tests. Here are their stories.

Wendy's choice

Wendy is 24 years old having her first baby. She wants as much information as possible on her pregnancy. She has decided to have one of the screening test options and knows that they have no risk to the baby. She has already had an ultrasound to check her dates and has decided to have the blood test at 15 weeks. Wendy understands that a screening test will give a risk estimate for Down syndrome that is more accurate than a risk based on her age. She knows that it is not a definite result. Wendy knows that low risk does not mean no risk as screening tests will miss some of the pregnancies that have Down syndrome. She also understands that an increased risk result is more likely to mean that the pregnancy is not affected by Down syndrome, but the only way to be sure is to have a diagnostic test. If there was an abnormality, Wendy doesn't think she would terminate the pregnancy. However, she thinks knowing about an abnormality before her baby is born would be helpful to her.

Tania's choice

Tania is a 28 year old woman in her first pregnancy. She has decided not to have any prenatal testing because she thinks that the risk of women her age is low for Down syndrome. She knows that screening tests will give her more information but doesn't think this will be of any use to her. She will have an ultrasound at 18-20 weeks and understands that this is a screening test for some conditions but feels that her risk of having a baby with any abnormalities is low.

Rebecca's choice

Rebecca is 37 years old having her third baby. She wants a test to give definite information so has decided to have a diagnostic test and not to have any screening tests. Rebecca is aware that this information only relates to chromosomal abnormalities and she will also have an ultrasound at 18-20 weeks to check the baby's structure. She has decided on the early diagnostic test (CVS), even though it has twice the miscarriage risk of the amniocentesis. Her decision is based on wanting to know as early as possible before she tells her family that she is pregnant. If there was an abnormality she doesn't think she would continue with the pregnancy.

Sue's choice

Sue is a 40 year old woman who is pregnant for the first time after trying to have a baby for years. She is aware of the increased risk of women her age for chromosomal abnormalities like Down syndrome but wants to avoid any risk of miscarriage. She has decided to have Combined First Trimester Screening. Sue understands that because of her age it is more likely the result will be “increased risk” but feels that if her risk is less than the risk of other women her age then she will probably not have a diagnostic test. Sue is also not sure what she would do if there was a problem.

What may happen to Wendy, Tania, Rebecca and Sue?

Tania will only have information on her age-related risk of having a baby with Down syndrome until she has her 18 – 20 week ultrasound. This

ultrasound may indicate that further testing, such as an amniocentesis, should be offered. If Wendy and Sue receive a low risk result from their screening tests, they will still have a small chance of having a baby with Down syndrome. If either woman did have an increased risk result it is more than likely that the baby will not have Down syndrome. However, they would be offered a diagnostic test. For women like Rebecca, having a diagnostic test means an increased risk of miscarriage but the test does give a definite result of whether or not the fetus has a chromosomal abnormality.

Any one of these women may have a miscarriage regardless of whether or not they had testing.

However, it is most likely that Wendy, Tania, Rebecca and Sue went on to have healthy babies, regardless of what they decided about testing.

Resources

Prenatal Diagnosis and Genetic Counselling Services

Genetic Health Services Victoria	8341 6303
Royal Women's Hospital	9344 2121
Monash Medical Centre	9594 2026
Mercy Hospital for Women	9270 2222

Web sites

Source: Department of Human Services Victoria <http://www.health.vic.gov.au/maternity/>
<http://www.betterhealth.vic.gov.au/>
Source: Commonwealth Government <http://www.healthinsite.gov.au/>
Cochrane Collaboration Consumer Information <http://www.update-software.com/Abstracts/COMMUNAbstractIndex.htm>

Genetic Health Services Victoria www.genetichealthvic.net.au/

Genetics Education Center, University of Kansas <http://www.kumc.edu/gec/>

Antenatal Results and Choices, UK <http://www.arc-uk.org/>

Ottawa Health Research Institute: Personal Decision Guide <http://www.ohri.ca/DecisionAid/>

Work Sheet

These are the suggested steps to assist you in making decisions about prenatal testing for fetal abnormalities:

- Read the information in the booklet as often as you need to
- Review the benefits and risks of the different tests
- Clarify your point of view by completing the following steps in the work sheet
- Talk about these issues to your partner, family and/or friends if you need to
- Discuss any issues and/or questions with your doctor or midwife or contact one of the services detailed in the resource section of this Decision Aid.

There are 6 steps to the work sheet. There are instructions to guide you through and to tell you which steps to skip if necessary.

Step 1

Ask your GP, midwife or obstetrician to calculate the risks for **all** women your age of having a pregnancy affected with Down syndrome at 10 weeks and at the expected date of delivery. You can see how the risk of a pregnancy affected by Down syndrome decreases as your pregnancy progresses.

It is also important to consider what the chance is of **not** having a pregnancy with Down syndrome. For example a risk for Down syndrome of 1 in 200, also means a chance that 199 of 200 pregnancies will **not** be affected.

- Work out the chance for all women your age of **not** having a pregnancy affected with Down syndrome.

The chance for all women my age of **not** having a pregnancy with Down syndrome is:

< > out of < > at the time of the early screening (10 weeks)

< > out of < > at the expected date of birth.

- Place a cross above the word that best describes what you feel is the risk of having a pregnancy affected with Down syndrome for women your age.



Very low Low Medium High Very high

Two examples of how to complete Step 2 of the work sheet are provided on Pages 18 and 19.

Have a look at the examples and then continue your work sheet on page 20.

Work sheet Example A – Step 2

Will screening tests give Tania useful information?

- Tania's story appears on page 15 of the booklet. To Tania, the risk of a woman her age of having a pregnancy affected with Down syndrome is low, although she knows it can happen. She sees no reason to have testing.
- She doesn't see any benefit in having a test that would only give a risk result and not definite information.

Next to each statement for having and not having a screening test, Tania placed a number in the corresponding box according to how important the statement was to her. She used the following guide:

0 = The statement is not at all important or not relevant, 1 = The statement is slightly important, 2 = The statement is important, 3 = The statement is very important

These statements indicate screening tests **will not** be useful.

She would not terminate this pregnancy.

1

She thinks her risk of having a pregnancy with a fetal abnormality is low.

3

Even though an "increased risk" result does not mean a definite fetal abnormality, this result would cause her far too much anxiety.

1

She would not consider a diagnostic test even if a screening test gave an "increased risk" result.

0

These statements indicate screening tests **will** be useful.

Knowing about an abnormality before her baby is born would help her to be prepared.

0

She wants the greatest chance of detecting Down syndrome without any added risk of miscarriage.

1

If her pregnancy is at "increased risk" for Down syndrome, she can be offered a diagnostic test.

0

A "low risk" result would be reassuring to her even though she knows this doesn't mean "no risk".

1

To decide whether or not screening tests would be useful to her, Tania looked at the column that had any numbers "2" & "3" marked in it, indicating that she considered the statement important. Tania has placed a "3" next to a statement that indicates screening would not be useful to her. She has then put a cross on the line below indicating that her leaning is towards not having a screening test.

For Tania, the information from a screening tests would be...

Not at all useful

Very useful

X

Work sheet Example B – Step 2

Will screening tests give Wendy useful information?

- Wendy's story appears on page 15 of the booklet. Wendy wants as much information as possible on her risk for having a pregnancy with Down syndrome. This is very important to her. She understands that the information from screening tests is limited.
- She does not want any added risk of miscarriage, but would have a diagnostic test if her result showed an "increased risk".

0 = The statement is not at all important or not relevant, 1 = The statement is slightly important, 2 = The statement is important, 3 = The statement is very important

These statements indicate screening tests **will not** be useful.

She would not terminate this pregnancy.

2

She thinks her risk of having a pregnancy with a fetal abnormality is low.

1

Even though an "increased risk" result does not mean a definite fetal abnormality, this result would cause her far too much anxiety.

0

She would not consider a diagnostic test even if a screening test gave an "increased risk" result.

0

These statements indicate screening tests **will** be useful.

Knowing about an abnormality before her baby is born would help her to be prepared.

3

She wants the greatest chance of detecting Down syndrome without any added risk of miscarriage.

2

If her pregnancy is at "increased risk" for Down syndrome, she can be offered a diagnostic test.

2

A "low risk" result would be reassuring to her even though she knows this doesn't mean "no risk".

1

Wendy has placed a "3" next to a statement that indicates screening would be useful to her. She has then put a cross on the line below indicating that her leaning is towards having a screening test.

For Wendy, the risk information from a screening tests would be...

Not at all useful

Very useful

X

The steps for you to complete continue on Page 20. Instructions to guide you through the steps are found at the bottom of the pages.

Your Work sheet

Step 2. Will screening tests give me useful information?

For **Steps 2 – 5** read the statements in both columns. Place a number in the corresponding box according to how important the statement is to you. Use the following guide:

0 = The statement is not at all important or not relevant, 1 = The statement is slightly important, 2 = The statement is important, 3 = The statement is very important

These statements indicate screening tests **will not** be useful.

I would not terminate this pregnancy.

I think my risk of having a pregnancy with a fetal abnormality is low.

Even though an "increased risk" result does not mean a definite fetal abnormality, this result would cause me far too much anxiety

I would not consider a diagnostic test even if a screening test gave an "increased risk" result.

These statements indicate screening tests **will** be useful.

Knowing about an abnormality before my baby is born would help me to be prepared.

I want the greatest chance of detecting Down syndrome without any added risk of miscarriage.

If my pregnancy is at " increased risk" for Down syndrome, I can be offered a diagnostic test.

A "low risk" result would be reassuring to me even though I know this does not mean there is no risk.

- Now look at the column that has any numbers "2" & "3" marked in it, indicating that you consider the statement important. (If there are none, then look for any number "1").
- Using this information now place a cross on the line below, to indicate the strength of your preference towards having or not having **a screening test**.

Not at all useful

Very useful

If you definitely prefer not having any testing at this point, go to Step 6.

If you are unsure or are leaning towards testing continue to Step 3.

Step 3. Which screening test?

One of the **earlier** screening tests, Combined First Trimester Screening or Nuchal Translucency, would be useful to me because..

I want a screening test with the highest detection rate.

I want my risk information as early as possible, before I tell others I am pregnant.

If I receive an "increased risk" result I want the earlier diagnostic test (CVS).

Earlier testing will give me the option of an earlier termination of pregnancy if a major abnormality is detected.

The **later** screening test, Maternal Serum Screen, would be useful to me because..

I also want to have risk information on neural tube defects like spina bifida.

If I receive an "increased risk" result I want the safer diagnostic test (amniocentesis).

It is free of charge for public patients.

I need more time to decide whether or not to have a screening test.

- Using this information now place a cross on the line below, to indicate the strength of your preference towards having or not having **a particular screening test**.

**I definitely prefer
Combined First
Trimester Screening**

**I definitely prefer
Second Trimester Maternal
Serum Screening**

If you have decided to have a screening test go to Step 6.

If you are unsure or want to consider diagnostic tests go to Step 4.

Step 4. A screening or a diagnostic test?

For women 37 years and older diagnostic tests are free of charge if you are a public patient.

0 = The statement is not at all important or not relevant, 1 = The statement is slightly important, 2 = The statement is important, 3 = The statement is very important

Information from **Screening tests** will be useful to me because...

It will give me a better chance of picking up a fetal abnormality than using my age risk alone.

If the result I receive is a risk less than other women my age this would be helpful to me in deciding whether or not to have a diagnostic test.

A "low risk" result would be reassuring to me even though I know this does not mean there is no risk.

I want more information on my individual risk before considering a diagnostic test because of the added miscarriage risk.

Information from **Diagnostic tests** will be useful to me because...

I want more information than a risk result.

I want the best chance to pick up chromosomal abnormalities.

I consider my risk of having a pregnancy with Down syndrome is high compared to my risk of miscarriage from a diagnostic procedure.

I would not continue with a pregnancy with Down syndrome.

- Using this information now place a cross on the line below, to indicate the strength of your preference towards having or not having **a screening or diagnostic test.**

I definitely prefer the features of screening tests.

I definitely prefer the features of diagnostic tests.

If you definitely prefer a screening test at this point, go to Step 6.

If you are unsure or are leaning towards diagnostic testing, continue to Step 5.

Step 5. Which diagnostic test?

For women 37 years and older diagnostic tests are free of charge if you are a public patient.

CVS, the earlier diagnostic procedure is useful to me because...

I want the earliest procedure available to exclude a chromosomal abnormality before I tell others of my pregnancy.

In the unlikely event of the diagnosis of a chromosomal abnormality I would want to avoid a termination of pregnancy involving an induced labour.

Amniocentesis, the later diagnostic procedure is useful to me because...

I want the safest diagnostic procedure.

The earlier test might pick up pregnancies with fetal abnormalities that were going to miscarry anyway. By waiting a few weeks I give nature a better chance to take its course.

- Using this information now place a cross on the line below, to indicate the strength of your preference towards having or not having **a particular diagnostic test**.

**I definitely prefer
the features
of CVS**

**I definitely prefer
the features
of amniocentesis**

Step 6. Weighing up Prenatal Testing options

Review your preferences for the relevant options relating to prenatal testing. Place a cross **X** on each line closest to your preference. Leave the options that are not relevant to you blank.

I consider that my age related risk for having a pregnancy with Down syndrome is...

Low **High**



For me, the information from a screening tests would be...

Not at all useful **Very useful**



Of the different screening tests, I would prefer to have...



Of the different types of tests I would prefer to have a ...



Of the different diagnostic tests I would prefer to have a/an...



Overall my decision about prenatal testing is

Summary of Tests

Screening tests are available to women of all ages. These tests give risk results not a diagnosis. Refer to pages 6-8.

	What is the test? When is it done?	What information will it give me?	How safe is it?
Combined First Trimester Screen	A blood test (between 8 and 12 weeks) and an ultrasound (nuchal translucency)	Detects up to 90% of pregnancies with Down syndrome, detects Edwards syndrome, confirms due date, identifies twins, identifies miscarriage	No added risk of miscarriage
Nuchal Translucency	An ultrasound between 11½ and 14 weeks An accredited provider of this test is recommended	Can detect pregnancies with Down syndrome, confirms due date, identifies twins, identifies miscarriage	No added risk of miscarriage
Second Trimester Maternal Serum Screen	A blood test between 14 and 20 weeks (Recommended between 15 and 17 weeks)	Detects up to 75–80% of pregnancies with Down syndrome Also gives a risk result for neural tube defects & Edwards syndrome	No added risk of miscarriage
Second Trimester Ultrasound	An ultrasound between 18 and 20 weeks	Detects many physical fetal abnormalities such as spina bifida Measures fetal growth and locates the position of the placenta	No added risk of miscarriage
Diagnostic tests for women at increased risk of fetal abnormality. Refer to pages 12-13.			
Chorionic Villus Sampling (CVS)	A sample of the developing placenta is taken using a fine needle guided by an ultrasound between 10 and 13 weeks	Provides a diagnosis of all major chromosomal abnormalities	1% (1 in 100) additional risk of miscarriage
Amniocentesis	A sample of fluid is taken from around the developing fetus using a fine needle guided by an ultrasound from 15 weeks	Provides a diagnosis of all major chromosomal abnormalities	0.5% (1 in 200) additional risk of miscarriage

Acknowledgments

This Decision Aid has been produced by a team of health professionals from:

Public Health Genetics Unit & Education Unit, Murdoch Childrens Research Institute, Melbourne, Victoria

Department of General Practice, University of Melbourne

Jean Hailes Foundation & Monash University Dept of Obstetrics and Gynaecology, Melbourne

Department of Medical Oncology, Prince of Wales Hospital, Randwick

Clinical Epidemiology and Biostatistics Unit, Royal Children's Hospital

The team have expertise in prenatal testing from their backgrounds in genetic counselling, obstetrics, midwifery, psychology, general practice, epidemiology and education.

The ADEPT project team gratefully acknowledges the valuable contributions from the many women and GPs who were vital to the development of this resource.

The development and evaluation of ADEPT was funded by the National Health and Medical Research Council (Project Grant no 237124).

© 2004 Murdoch Childrens Research Institute

Design by bluemoon design

