



Prenatal Testing

**Special tests for your
baby during pregnancy**

Includes Reproductive
Carrier Screening

January 2024



Centre for Genetics Education

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NSW Health's Centre for Genetics Education aims to raise awareness of the role that genetics plays in family health, both for health professionals and the public.

The Centre can be contacted regarding the availability of a range of genetic services, information and resources. Information about genetics clinics and genetic support groups is also available.

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Further copies of this booklet are available from the Centre or can be downloaded from the Centre's website www.genetics.edu.au

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ABOUT THIS BOOKLET

This booklet is for individuals and couples who are planning a pregnancy or who are already pregnant and want further information about the tests available before and during pregnancy. Tests during pregnancy are called prenatal tests.

Everyone wants to have a healthy baby. However, there are some people whose baby may have or will develop a serious physical and/or intellectual condition.

There are different tests and procedures available to assess the health and development of a baby before birth. Each has advantages, disadvantages and limitations. The decision to undergo testing during a pregnancy is a very personal one and a decision best made based on all the available information.

In some places this booklet uses terminology such as woman, mother and female to describe people experiencing pregnancy and father, man or male to describe people who produce sperm. We recognise that not everyone will identify with these terms. Our intention is to be inclusive of all people who can experience and contribute to pregnancy.

It is important to remember that you do not have to have prenatal testing if you do not wish to.

HOW THIS BOOKLET CAN HELP YOU

This booklet provides information about the different types of prenatal tests available during different stages of pregnancy. You might find that there are similar tests that will provide the same information, but they are offered at different stages of pregnancy. For this reason, the booklet is set out to follow a simple timeline of pregnancy that you will see on page 6.

Screening tests during the pregnancy can provide information about the chance that a baby has a particular condition. Diagnostic tests during the pregnancy can provide more certain information about whether the baby has a particular condition. It is important to know the difference between a screening test and a diagnostic test. If a screening test shows an increased chance for a baby having a condition, then the person can decide if they wish to follow up with a diagnostic test for a certain result.

A glossary is also provided (see pages 41-47) to explain words used that might not be familiar to you.

Since some prenatal tests are specialised, they might not be available at all medical facilities. If you are planning a pregnancy or are already pregnant, contact your family doctor or one of the prenatal testing services listed on pages 37 to find out what is available in your area.

WHAT INFORMATION DOES A PRENATAL TEST PROVIDE?

A prenatal test is usually done to determine if your baby is developing in the usual way or if there is a chance that the baby could have a specific condition. Most of these tests can be done during pregnancy. One test (called reproductive carrier screening) can be done both during and before pregnancy. If possible, it is recommended to do this before pregnancy.

Some of the conditions that prenatal tests are able to detect include:

1. Genetic conditions

Genetic conditions include many of the health or developmental problems seen at birth, in childhood, adolescence or adulthood. A genetic condition can occur for a number of reasons. However, the types of genetic conditions that prenatal tests can identify could be caused by a chromosome imbalance (chromosome condition) or by a change in a single gene.

A chromosome condition occurs when a baby has a change in the number, size or structure of their chromosomes. This change in the amount or arrangement of the genetic information in the cells may result in problems in growth, development and/or functioning of the body systems.

Chromosome changes can be inherited from a parent. More commonly, chromosome changes occur when the egg or sperm cells are forming, during conception or just after. The reason for this change is unknown (spontaneous occurrence). The most commonly known chromosome condition is Down syndrome.

Other genetic conditions, which are caused by a variation in a single gene, are known to affect babies in some families. This could be because the couple have already had a child with one of these conditions or because one of the parents has the condition themselves. In these situations, a prenatal test can be used to specifically identify if the baby has this genetic condition. (see Glossary)

2. Neural tube defects

Neural tube defects occur when part of the spine or brain is not formed correctly. One such condition is spina bifida (meaning open spine). (see Glossary, page 47)

A prenatal test cannot check every possible physical, developmental or intellectual condition that can affect your baby.

WHAT ARE PRENATAL TESTS?

Prenatal tests can assess the health and development of your baby during pregnancy. There are two main types of prenatal tests:

1. Prenatal screening tests

Screening tests cannot tell if your baby definitely has a genetic condition but they might indicate that further testing needs to be considered (diagnostic testing). Regardless of your age or family health history, you can choose whether or not you have a prenatal screening test.

Prenatal screening tests include:

- Reproductive carrier screening (recommended before falling pregnant)
- Ultrasound
- Non-invasive prenatal testing (NIPT): testing of the mother's blood
- First trimester (early pregnancy) screening: nuchal (pronounced new-cal) translucency ultrasound with or without testing of the mother's blood
- Second trimester (mid pregnancy) screening: testing of the mother's blood (maternal serum testing)

2. Prenatal diagnostic tests

Diagnostic tests look for a specific condition that your baby might be at risk of. These types of tests can assess your baby for a chromosome condition or a condition caused by a variation in a single gene. A diagnostic test does not check every possible physical or intellectual problem that could affect your baby.

Prenatal diagnostic tests include:

- Ultrasound
- Chorionic villus sampling (CVS)
- Amniocentesis (pronounced am-nee-o-cen-tee-sis)

Each prenatal test is done at a certain time during the pregnancy (see Prenatal testing timeline on page 6)

THINGS TO CONSIDER WHEN MAKING A DECISION ABOUT PRENATAL TESTING?

Pregnancy is usually divided into three stages (trimesters). The first trimester refers to the first three months of pregnancy, the second trimester spans months four to six and the third trimester covers the final three months of pregnancy. Prenatal tests are done in either the first or second trimester of pregnancy.

It is strongly recommended that you get as much information as possible before any testing is done. Discussions regarding prenatal testing options should be offered to you and your partner in a safe and understanding environment. Seeking support at this time can assist you in making informed decisions about the future of your pregnancy.

It is important to discuss the following prenatal testing issues:

- How and when the tests are done
- The advantages and disadvantages of each test
- Any risks to you or your baby that may result from each test
- Any further testing that might be offered and what it involves.

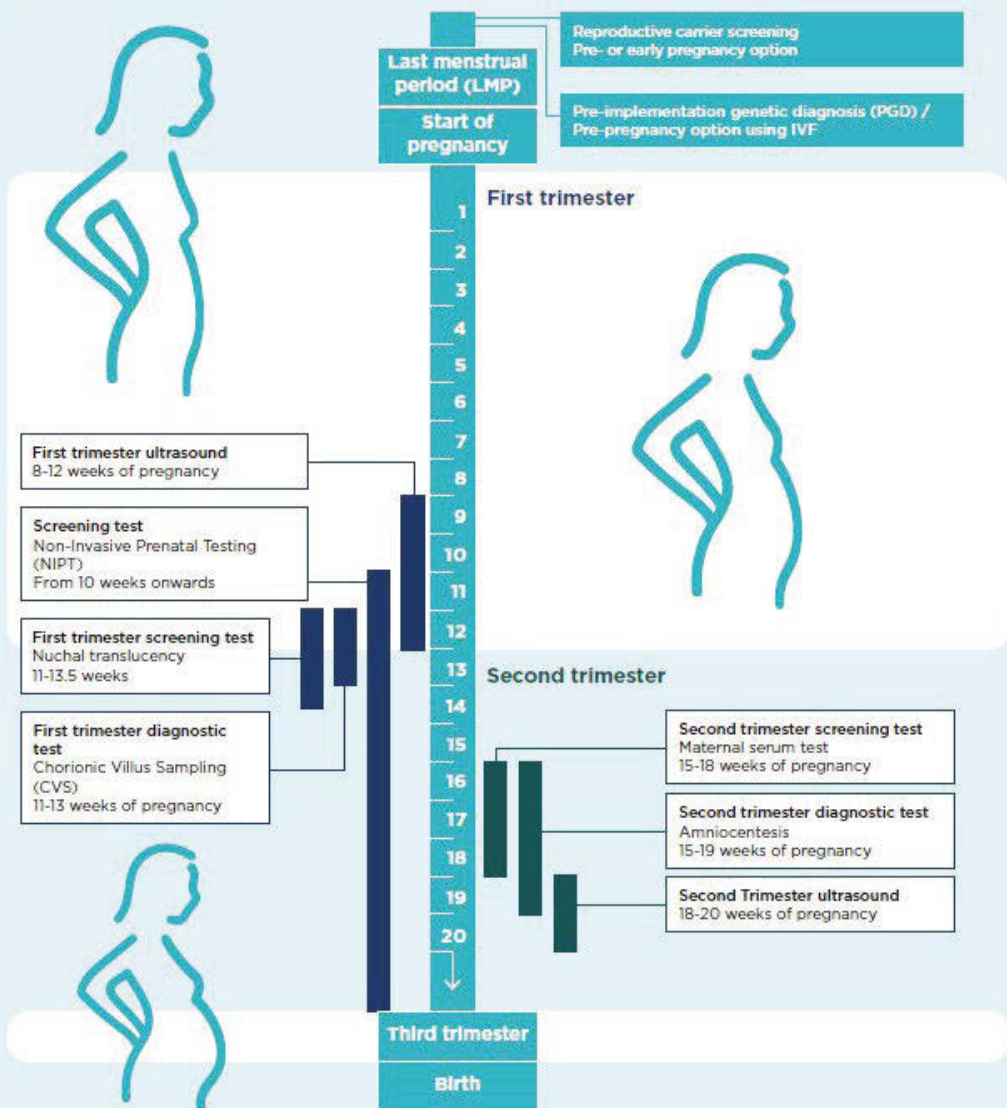
If the result of a prenatal test shows that your baby is not developing normally or could develop a problem after birth, genetic counselling will give you the opportunity to discuss:

- What the result means for your baby and your family
- The options available at this time such as further testing and what it involves
- Your thoughts and feelings towards potentially having a child with a disability and medical needs (note: the different conditions vary a lot. Your health care provider should give you specific information about the condition relevant to your baby)
- Your thoughts and feelings towards termination of pregnancy.

A list of services where you can find out about the availability of prenatal testing and genetic counselling in your area is available on pages 37-39.

Counselling before a test is done, will help you decide which test, if any, is best for you and your baby.

Prenatal Testing Timeline



NOTES ABOUT MY PREGNANCY

Date of the first day of my last menstrual period (LMP):

My expected date of delivery (EDD):

Appointments:

Contacts:

Notes:

Reproductive Carrier Screening

Prior to pregnancy or in the first 12 weeks of pregnancy

- The test can identify a couple's chance of having a child with specific genetic conditions
- Does not pose any health risk to you or your baby
- As with all tests during pregnancy, reproductive carrier screening is optional

What is Reproductive Carrier Screening (RCS)?

RCS is a blood or saliva test that looks at a person's genes. RCS can determine if one or both parents are genetic carrier(s) of some recessive or X-linked genetic condition(s) (see pages 46-48). These genetic conditions can affect the health and development of any children they may have together.

How is RCS done?

RCS can test for a number of different genetic conditions at one time. The test requires a blood or saliva sample from one or both biological parents. Usually, the tests are carried out in one of the following ways:

- **A 3-gene carrier screen** which tests for three common genetic conditions that have a serious impact on the health of an affected individual: cystic fibrosis (CF), spinal muscular atrophy (SMA) and fragile X syndrome. Approximately 1 in 20 people in the Australian population are a genetic carrier for one of these three conditions and the majority of couples "with an increased chance" do not have a family history of the condition. This test will be covered by Medicare from November 2023.
- **Expanded carrier screening** can test for many different genetic conditions, sometimes tens or hundreds, most of which are rare. The expanded carrier screening test will identify if someone is a genetic carrier of the 3 common conditions CF, SMA and fragile X syndrome, as well as many others that are severe and can start early in life. This test is not covered by Medicare.

How accurate is Reproductive Carrier Screening?

Reproductive carrier screening is a screening test. It will provide information about the chance that your baby will have a certain genetic condition. Couples will receive an increased chance or low chance result.

- An increased chance result means that one parent carries an X-linked condition or both parents carry the same recessive genetic condition. When both parents carry the same recessive condition, there is a 25% (1 in 4) chance of having a baby with that condition. When a parent carries an X-linked condition the chance will depend on the condition and the result
- A low chance result means that the test did not identify any X-linked conditions and the parents are not carriers of the same recessive genetic conditions. This does not guarantee a healthy baby but it means there is a very low chance of having a child with any of the genetic conditions you were screened for.

What if Reproductive Carrier Screening shows that you have a higher chance of having a baby with a genetic condition?

If you receive an increased chance result, how you use this information is completely up to you. You should discuss your options with your healthcare provider and take into consideration your personal preferences. Some of the options are discussed in more detail in this booklet and include:

- Falling pregnant naturally (or continuing if you are already pregnant) and having no extra testing for the genetic condition in the pregnancy
- Falling pregnant naturally and having a Chorionic Villus Sampling (CVS) (see pages 18-23) or Amniocentesis (see pages 27-30) to see if the pregnancy has the genetic condition. If the pregnancy has the condition you would have the option to end the pregnancy (termination)
- Falling pregnant using Pre-Implantation Genetic Diagnosis (PGD) (see pages 35-36) to reduce the chance of having a child with the genetic condition
- Falling pregnant using In Vitro Fertilization (IVF) and using a sperm or egg donor to reduce the chance of having a child with the genetic condition

First trimester ultrasound

8-12 weeks of pregnancy

- An important step if you are considering having testing later in your pregnancy
- A safe way of assessing the growth of your baby in the first trimester of pregnancy
- Does not pose any health risk to you or your baby
- First trimester ultrasounds, like all other prenatal tests, are optional

What is first trimester ultrasound?

Ultrasound uses harmless high frequency sound waves to produce images of your developing baby in the uterus (womb).

During an ultrasound, a transducer (that acts like a microphone) is used to generate sound waves that produce images on a screen. The transducer can do this either through the mother's abdomen or vagina. Your doctor or sonographer (specially trained ultrasound technician) will discuss which procedure is best for you.

During an **abdominal ultrasound**, the doctor or sonographer will use a water-based jelly-like substance and move the transducer over your abdomen. Sound waves pass from the transducer into the uterus and through the fluid surrounding the baby (called amniotic fluid). The sound waves bounce harmlessly off the baby, creating echoes.

For abdominal ultrasound to be most successful, a full bladder is needed. The uterus and ovaries are often hidden behind the bowel, making it difficult to clearly see inside the uterus. However, when the bladder is full the bowel is pushed out of the way. It is recommended that you have several glasses of non-fizzy fluid one hour before the ultrasound test.

During a **vaginal ultrasound**, the transducer is passed into the vagina using sterile techniques. The sound waves pass into the uterus to create the same echoes as described above. For vaginal ultrasound it is best if the bladder is empty.

Why should I consider having a first trimester ultrasound?

The most common reasons for having a first trimester ultrasound are to:

- Check or confirm how many weeks the pregnancy has progressed
- See if there is more than one baby in the uterus
- View the position of the baby and the placenta
- Check on the wellbeing of the baby when there has been a complication such as bleeding.

A first trimester ultrasound may also be referred to as a dating ultrasound.

When is the first trimester ultrasound done?

An ultrasound scan can be done at any time during the pregnancy. The best time for determining the date and stage of pregnancy, and the number of babies in the uterus is between 8 and 12 weeks.

When will the result of the first trimester ultrasound be available?

In some cases, information about your baby's ultrasound scan will be made available immediately. However, if the person doing the scan suspects that your baby has a problem, he or she may not be able to discuss the findings until they have been verified by another specialist. The result might be sent to the referring doctor who can discuss the result with you.

It might be useful to discuss how the result will be given, to whom and when, with the sonographer or doctor before the ultrasound.

What will the result of the first trimester ultrasound tell me?

The first trimester ultrasound will provide accurate dating of a pregnancy which is often needed if a woman is considering having a prenatal test later in the pregnancy.

How reliable is the first trimester ultrasound?

A first trimester ultrasound cannot check for every possible physical, developmental or intellectual problem that could affect your baby. First trimester ultrasound is very reliable at determining how advanced the pregnancy is and how many babies there are.

What if the result of the first trimester ultrasound shows my baby has a problem?

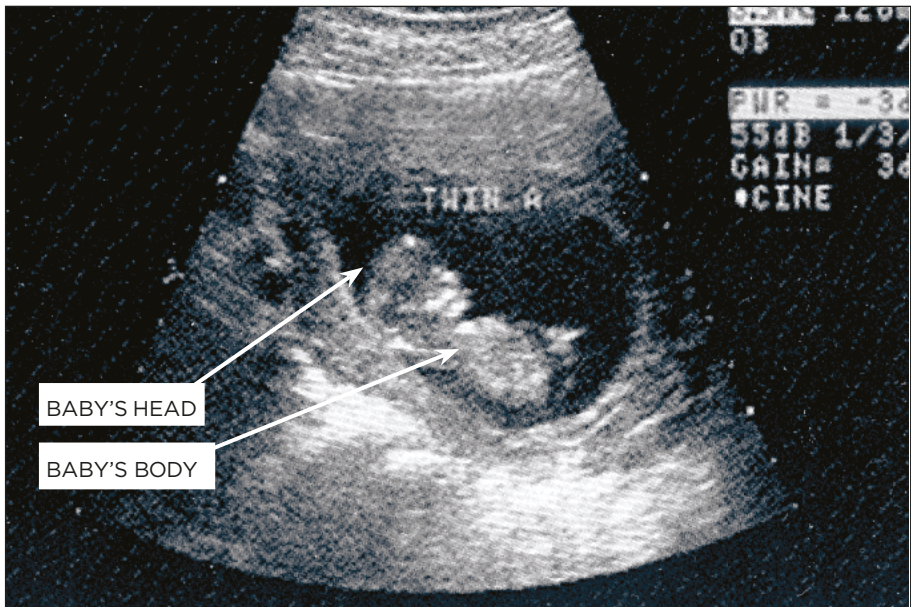
In a small number of cases, an ultrasound done early in the pregnancy might show that the baby either has or is at risk of having a problem. A more specialised ultrasound scan to determine whether or not the baby actually has a problem might be suggested. Alternatively, depending on how advanced the pregnancy is, the doctor might also discuss the option of having one of the diagnostic prenatal tests (see CVS and amniocentesis, pages 18-30).

Can the first trimester ultrasound harm me or my baby?

No evidence has been found that shows an ultrasound can harm you or your baby.

Discussing your result and options with your doctor, midwife or genetic counsellor will help you to decide if you want further testing and if so, which test is best for you.

ULTRASOUND SCAN AT 11.5 WEEKS



Screening Test - non-invasive prenatal testing (NIPT)

10 weeks onwards

- Screening test for Down syndrome and some other chromosome conditions
- Involves a special blood test
- Provides an accurate estimate of whether or not your baby has Down syndrome or some other chromosome conditions
- Does not pose any health risk to you or your baby
- NIPT is a new test - currently only offered through some specialist centres
- NIPT, like all other prenatal tests, is optional

What is non-invasive prenatal testing?

Non-invasive prenatal testing (NIPT) is a screening test used to determine the likelihood of your baby having Down syndrome (see Glossary, pages 41-47) or some other chromosome conditions.

The test can be used from 10 weeks of pregnancy and requires a sample of the mother's blood. During pregnancy, some of the DNA from the baby (called foetal DNA) crosses into the mother's bloodstream. This foetal DNA carries the baby's genetic information. It is this foetal DNA in the mother's blood that is analysed.

Why should I consider having non-invasive prenatal testing?

This test is highly accurate for detecting Down syndrome. It is also effective at detecting some other chromosome conditions. NIPT is a safe way to screen for these conditions during your pregnancy.

When is non-invasive prenatal testing done?

The test can be used as early as 10 weeks of pregnancy.

When will the result of non-invasive prenatal testing be available?

In most circumstances it will take 2 weeks for you to receive a result. In a small number of cases the lab is not able to provide a result and if this occurs, your doctor, midwife or genetic counsellor can discuss alternative testing options with you.

What will the result of non-invasive prenatal testing tell me?

A negative, normal or low risk result indicates that the baby is unlikely to be affected by any of the chromosome conditions included in the screen. A positive, abnormal or high risk result indicates that the baby is likely to be affected by the specified chromosome condition.

The risk result you receive is based on factors in your current pregnancy only. This risk is worked out based on the foetal DNA obtained from the mother's blood.

Find out from your doctor, midwife or genetic counsellor exactly which chromosome conditions the testing service they use will be looking for. As NIPT becomes more available, there may be new health conditions being included in the screening of the foetal DNA.

How reliable is non-invasive prenatal testing?

As with all screening tests, the results of NIPT will not give a 100% definite answer about whether or not your baby has a chromosome condition. NIPT has been shown to be highly accurate in identifying babies with Down syndrome. The effectiveness may depend on the way in which the testing is done and where. Your doctor, midwife or genetic counsellor can inform you about the reliability of the test for Down syndrome and other chromosome conditions.

What if the result of the NIPT shows my baby might have a problem?

If the NIPT result shows your baby might have a chromosome condition, then you may want to consider having a diagnostic test like chorionic villus sampling (CVS) (see pages 18-23) or amniocentesis (see pages 27-30) to confirm the result. Your doctor, midwife or genetic counsellor can discuss these options with you.

Can the NIPT harm me or my baby?

The test is conducted using a sample of the mother's blood and does not pose any health risk to the mother or the baby.

First trimester screening test - nuchal translucency

11-13.5 weeks of pregnancy

- Screening test for Down syndrome and some other chromosome conditions
- Involves a specialised ultrasound with or without a blood test
- Estimates the chance of your baby having a chromosome condition such as Down syndrome
- Identifies 75-90% of babies with Down syndrome
- Does not pose any health risk to you or your baby
- First trimester screening tests, like all other prenatal tests, are optional

What is first trimester screening?

This screening test is used to determine the likelihood of your baby having Down Syndrome (see Glossary, pages 41-47) and some other chromosome conditions.

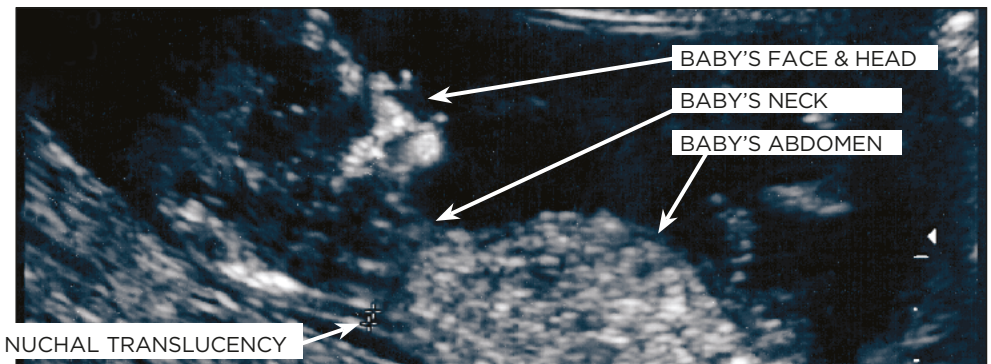
Between 11 and 13.5 weeks a specialised ultrasound will be used to measure your baby's nuchal translucency (a fluid-filled space at the back of your baby's neck).

At this early stage of the pregnancy, your baby's skin is very thin and fluid can be seen in the space between the skin and tissue at the back of the neck. The depth of the fluid in this space can be measured using ultrasound.

An additional blood test might also be done but is not always offered or available. This special blood test is not offered on its own at present, but only combined with the nuchal translucency ultrasound.

First trimester screening is also called nuchal translucency screening (NT screening or NT Plus when it includes the blood test).

This is what a nuchal translucency ultrasound picture looks like.



Why should I consider having first trimester screening?

First trimester screening is considered to be a safe way that a woman of any age can screen a pregnancy in the first trimester for certain chromosome conditions.

When is first trimester screening done?

The measurement of nuchal translucency using ultrasound is done between 11 and 13.5 weeks of pregnancy. Only doctors or sonographers with special training should take this measurement (see Services: Prenatal testing on page 37).

If you are also having the special blood test, it can be done before or at the same time as the nuchal translucency ultrasound. The blood test will measure the levels of specific proteins in your blood and when used in combination with the nuchal translucency ultrasound result, improves the accuracy of this screening test. It is important that the blood test result is calculated using the correct weeks of pregnancy and accurate maternal weight.

When will the result of first trimester screening be available?

If you are having the nuchal translucency ultrasound without the blood test, you may be given a result immediately.

If first trimester screening includes a blood test, the result will usually be available one week later.

What will the result of first trimester screening tell me?

The result of the first trimester screening test will be given to you in the form of a risk number (such as a chance of 1 in 100 or 1 in 500) that the baby has a chromosome condition.

The risk result you receive will be your own individually calculated risk based on factors in this individual pregnancy only. This number is worked out by a computer program using the thickness of the nuchal translucency and several other factors including maternal age and stage of pregnancy. If you have also had the special blood test, this is taken into account and provides a more accurate estimate.

A risk of 1 in 300 or greater (e.g. 1 in 150) is considered to be in the “increased risk” category.

You can discuss your result with your doctor, midwife or genetic counsellor and decide if you would like to have further testing in this pregnancy.

How reliable is first trimester screening?

This is a screening test and therefore will not give a definite answer about whether or not your baby has a chromosome condition.

Nuchal translucency alone: If you have the nuchal translucency ultrasound on its own, the test is around 75% accurate at identifying babies that have a chromosome condition like Down syndrome and they will receive a result in the “increased risk” category. Therefore, around 25% of babies with a chromosome condition will not be identified using nuchal translucency.

Nuchal translucency plus the special blood test: The nuchal translucency ultrasound together with the special blood test is around 80 to 90% accurate at detecting babies that have a chromosome condition like Down syndrome and they will receive a result in the “increased risk” category. Therefore, around 10 to 20% of babies with a chromosome condition will not be identified using this test.

It is important to note that an increased risk result does not mean your baby will definitely have a chromosome condition. Likewise, if your result is not in the increased risk category, your baby could still have a chromosome or other health condition.

What if the result of first trimester screening shows my baby has a problem?

About 5% of women (1 in 20) who have this test will receive an increased risk result. However, most of these babies will not have a chromosome condition.

If the result of first trimester screening suggests your baby is at an increased risk of having a chromosome condition or if you would like to have further testing, you may consider having either chorionic villus sampling (CVS) (see pages 18-23) or an amniocentesis (see pages 27-30). Any concerns you have should be discussed with your doctor, midwife or genetic counsellor.

Can first trimester screening harm me or my baby?

The first trimester screening test does not pose any health risk to you or your baby.

First trimester diagnostic test - chorionic villus sampling (CVS)

11-13 weeks of pregnancy

- Diagnostic test which gives an accurate result
- A sample of the chorion (placenta) is collected and tested to determine if the baby has certain genetic conditions
- Less than 1% of women (1 in 100) will have a miscarriage as a result of having a CVS
- CVS testing, like all other prenatal tests, is optional

What is chorionic villus sampling (CVS)?

Chorionic villus sampling (CVS) is a diagnostic test that uses a sample of the chorion (which develops into the placenta) to determine if your baby has certain genetic conditions. The cells that make up the chorion are mostly the same as the cells of your baby as they both originated from the developing embryo in early pregnancy.

There are two different methods that can be used to test the chorion. The sample can be collected either through the mother's abdomen (transabdominal) or the vagina (transvaginal). The most appropriate and safest method will depend on the position of the baby and the placenta and will be discussed with you before the procedure.

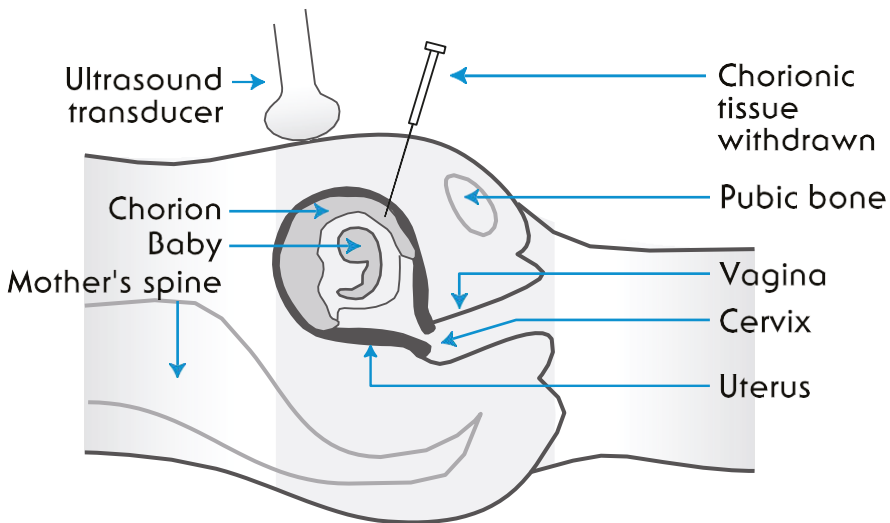
Transabdominal CVS uses a fine needle passed through your abdomen to obtain a sample of chorion. A local anaesthetic may be used on your skin prior to the procedure to numb the area where the needle is inserted. During the procedure, ultrasound is used to guide the specialist and enable the test to be carried out in the safest way.

The picture below shows a side-on view of a transabdominal CVS.

Using a tiny sample of the chorion, the test will usually examine your baby's chromosomes unless your pregnancy is at risk of another type of genetic condition (See Genetic Conditions in the Glossary, page 43).

An ultrasound scan is done before and during the test to enable the specialist performing the test to locate the position of the chorion. It also allows them to watch the sampling on the screen and makes sure the procedure is carried out as safely as possible.

The amount of chorion needed for the test is extremely small. Only about 0.001% (1 in 1000) of the total chorion is sampled during this test.

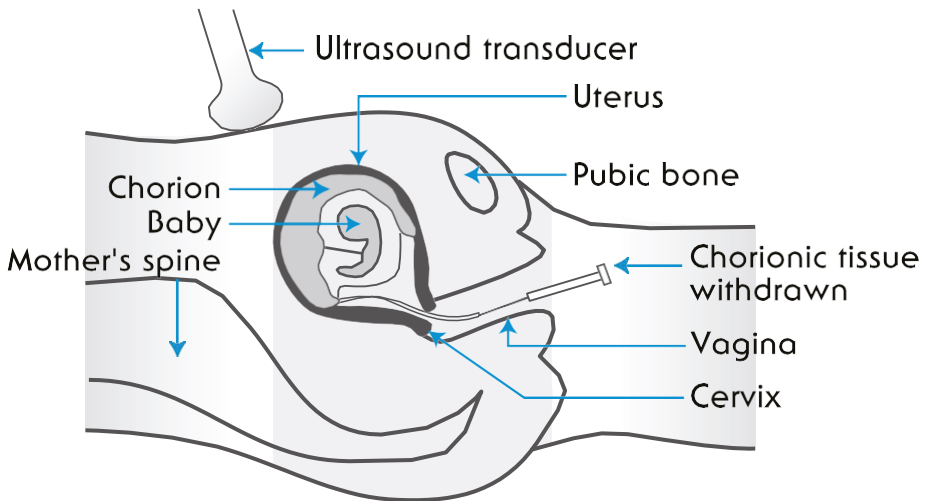


Source: After Vogel, F. & Motulsky, A.G. (1986). *Human Genetics, 2nd edn.* Springer-Verlag, Berlin.

The picture below shows a side-on of a transvaginal CVS.

During a transvaginal CVS, an ultrasound is used to locate the developing baby in the uterus. An instrument called a speculum is used to widen the opening of the vagina so that the cervix can be clearly seen by the doctor. The cervix is swabbed to remove mucus that is usually present and a fine tube is passed through the cervix into the uterus.

The image of the tube can be seen in the ultrasound picture and thus it can be guided to the chorion and remove a sample for testing. The picture below shows a side-on view of this.



Source: After Vogel, F. & Motulsky, A.G. (1986). *Human Genetics, 2nd ed.* Springer-Verlag, Berlin.

Why should I consider having a CVS?

- You received an increased risk result on non-invasive prenatal testing (NIPT) (see pages 13-14)
- You received an increased risk result on a first trimester screening test (see pages 15-17)
- You are over 35 years old. The risk of having a baby with a chromosome condition increases with the mother's age (see "Chromosome condition" in the Glossary, pages 41-47).
- You or your partner are known to be at risk of passing on a chromosome condition or other type of genetic condition caused by a gene variation. When the CVS is done to check for a gene variation, the laboratory will need to establish a gene test which can then be used during the CVS. This type of testing is very specialised and usually organised by a genetic counsellor.

When is a CVS done?

A CVS is usually done between 11 and 13 weeks of pregnancy. In special circumstances, the CVS might be carried out later in your pregnancy. However, it should not be done before 10 weeks of pregnancy.

When will the result of a CVS be available?

It might take two to three weeks to receive your CVS result as it can be a lengthy process. After a sample of chorion has been collected from you, it is sent to a specialised laboratory where the testing is done. The process involves growing the cells in a sterile environment and is, therefore, a time-consuming method. If the test is looking at the chromosomes, the laboratory may issue an initial result within two to three days. This will provide a result for the common chromosome conditions only. The final result will still take two to three weeks.

What will the result of a CVS tell me?

It is important to understand what information the test will identify. Depending on why you are having the test, the laboratory can focus on specific conditions e.g. if you are having the test after receiving an increased first trimester screening risk result, then it will usually be a chromosome condition that is the concern.

For other women, they may have previously had a child with a specific genetic condition caused by a gene variation or a specific chromosome change. In this case, the laboratory would analyse that particular gene or look carefully at the chromosome in question in the developing baby.

Although doctors and scientists are now able to diagnose a large number of conditions during pregnancy, it is not possible to test for all of the problems that could affect a developing baby. Your doctor or genetic counsellor will discuss with you the range of conditions that can be identified using a CVS test and what each of these might mean for you and your baby.

It is important to remember that a normal test result does not exclude every possible problem with the baby.

How reliable is a CVS?

The CVS test is very accurate at identifying the conditions your baby is being tested for.

However, as the test is done early in pregnancy the chorion, which develops into the placenta, is immature. Occasionally, the doctor might be unable to obtain enough chorion on the first attempt. Samples of the chorion will be examined immediately to make certain there is enough chorion for the laboratory to work with. If not, another sample may need to be taken at this time.

In rare cases, the laboratory might be unable to get a definite answer and a second CVS will have to be done at a later time. In other cases, there will be a need for an amniocentesis to be done to check the CVS result. This means that a very small number of women may need to have both a CVS and an amniocentesis in the same pregnancy. For further information about amniocentesis (see pages 27-30).

What if the result of the CVS shows my baby has a problem?

If a CVS result shows that your baby has a problem, you and your partner will be given as much information as possible about the condition. This includes the implications it might have for the future health or development of the baby. You will be given time to make an informed choice about whether or not you would like to continue the pregnancy.

If you and your partner decide to continue the pregnancy following the diagnosis of a health or developmental problem with your baby, you will both be offered support and information before, during and after the birth of the baby.

If you decide to terminate the pregnancy, the procedure might require a general anaesthetic and/or hospitalisation. The method of termination will vary depending on how advanced the pregnancy is and the policy of the particular hospital or clinic. All aspects should be fully discussed with your doctor or genetic counsellor. You will be offered support and information before, during and after your hospital stay.

Please see the services, support and information section on pages 37-40 of this booklet for more details.

Can a CVS harm me or my baby?

The increase in risk of miscarriage due to having a CVS is < 1% (1 in 100 pregnancies tested with a CVS). This risk is in addition to your “background risk” of miscarriage that all women have in early pregnancy due to natural causes. It is important to discuss your background risk of miscarriage with your doctor, midwife or genetic counsellor.

The specific risk figure for a miscarriage depends on the experience of the doctor doing the test and the difficulty he or she has in obtaining the sample of chorion. It is therefore important that a CVS test is only carried out by a doctor experienced in this technique and should be done after the 11th week of pregnancy (see the Services: Prenatal testing, page 37).

Some women will experience cramping and occasionally some vaginal bleeding after a CVS. Talk to your doctor about what you can expect after the testing and what symptoms to look out for.

Second trimester screening test - maternal serum testing

15-18 weeks of pregnancy

- Screening test for Down syndrome and neural tube defects
- Involves having a blood test
- Provides you with an estimate of whether or not your baby has a chromosome condition such as Down syndrome
- Identifies 60-75% of babies with Down syndrome
- Identifies 95-100% of babies with a neural tube defect

What is second trimester screening?

Second trimester screening (also known as the maternal serum triple test or maternal serum quadruple test, depending on the number of hormones measured) is used to determine the likelihood of your baby having a chromosome condition such as Down syndrome or a neural tube defect such as spina bifida (see Glossary, pages 41-47).

It can only estimate the chance of your baby having a chromosome condition or neural tube defect, not provide a definite answer.

Second trimester screening uses a specialised blood test to measure the levels of several proteins in your blood (maternal serum). The levels of these proteins, combined with other factors such as your age, can provide an estimate of the likelihood that your baby has Down syndrome or a neural tube defect such as spina bifida.

Why should I have second trimester screening?

Second trimester screening is considered to be a safe way to screen a pregnancy in the second trimester for certain chromosome conditions and neural tube defects. Women may have either a first trimester screening test or second trimester screening depending on their gestation. It is usually not recommended for women to have both first trimester and second trimester screening.

When is second trimester screening done?

The second trimester screening test involves having a blood test between 15 and 18 weeks of pregnancy.

When will the result of second trimester screening be available?

The results will usually be available one week following blood collection. At this time, you will have the opportunity to discuss them with your doctor, midwife or genetic counsellor and decide if you would like to have further testing in your pregnancy.

What will the result of second trimester screening tell me?

The second trimester screening test provides you with an estimated chance of your baby having a chromosome condition such as Down syndrome or a neural tube defect such as spina bifida in this pregnancy. The result will be given to you in the form of a number such as a chance of 1 in 100 or 1 in 500 that the baby has one of these conditions.

A risk of 1 in 250 or greater (e.g. 1 in 100) is considered to be in the “increased risk” category.

This number is worked out by a specialist testing laboratory using the result of the blood test as well as other factors such as your age and stage of pregnancy.

How reliable is second trimester screening?

This is a screening test and therefore will not give a definite answer about whether or not your baby has a chromosome condition.

The second trimester screening test is around 60-75% accurate at identifying babies that have a chromosome condition like Down syndrome, depending on the number of pregnancy hormones measured. They will receive a result in the “increased risk” category. Therefore, around 25-40% of babies with a chromosome condition will not be identified using this test.

It is important to note that an increased risk result does not mean your baby will definitely have a chromosome condition. Likewise, if your result is not in the increased risk category, your baby could still have a chromosome or other health condition.

Most babies who have a neural tube defect will be identified using second trimester screening. If the screening result is used together with a second trimester ultrasound (see pages 31-34), the detection rate for some neural tube defects can be as high as 95-100%.

What if the result of second trimester screening shows my baby has a problem?

About 5% of women (1 in 20) who have this test will receive an increased risk result. It is important to note that most of these babies will not have a chromosome condition.

If the result of second trimester screening suggests your baby is at an increased risk of having a chromosome condition, or if you would like to have further testing to see if your baby has a chromosome condition, you might want to consider having an amniocentesis (see pages 27-30). Any concerns you have should be discussed with your doctor, midwife or genetic counsellor.

If the result suggests that your baby has a neural tube defect, you might want to have a second trimester ultrasound to view the developing baby and in particular the spine. See second trimester ultrasound on pages 31-34.

Can second trimester screening harm me or my baby?

The second trimester screening test does not pose any health risk to you or your baby.

Second trimester diagnostic test - amniocentesis

15-19 weeks of pregnancy

- Diagnostic test which gives an accurate result
- A sample of the amniotic fluid surrounding the baby is collected and tested to determine if the baby has certain genetic conditions
- Less than 1% of women (1 in 100) will have a miscarriage as a result of having an amniocentesis
- An amniocentesis, like all other prenatal tests, is optional

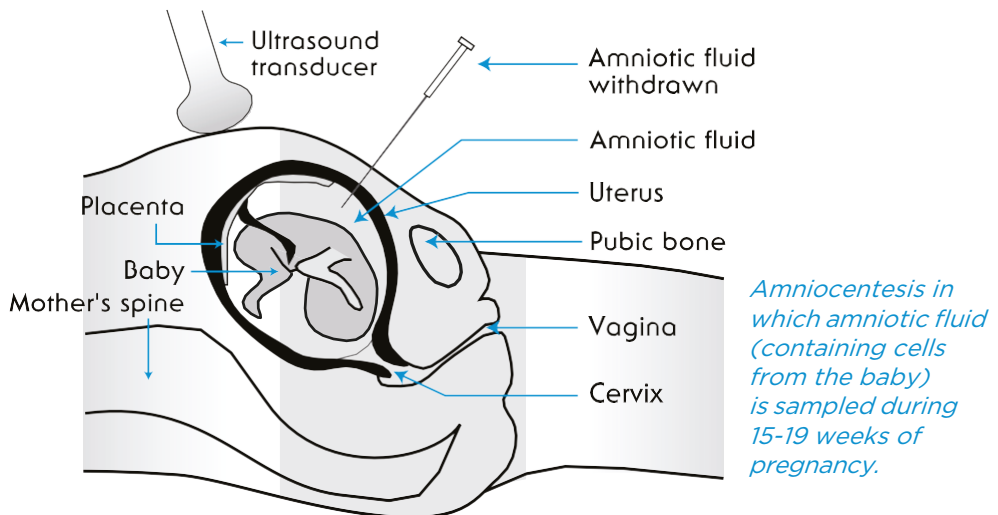
What is an amniocentesis?

An amniocentesis is a diagnostic test that will use a sample of the amniotic fluid that surrounds the baby to determine if the baby has particular genetic conditions.

Using a sample of the amniotic fluid, which contains cells from the baby, the test will usually examine your baby's chromosomes unless your pregnancy is at risk of another type of genetic condition (See Glossary, pages 41-47).

During an amniocentesis, a fine needle will be inserted into your abdomen to obtain a sample of amniotic fluid. A local anaesthetic might be used on your skin to numb the area where the needle is going to be inserted. The picture below shows a side-on view of this.

An ultrasound scan is done before and during the test, enabling the doctor to locate the position of the baby. It also allows them to watch the procedure on the screen and make sure it is carried out as safely as possible.



Source: After Vogel, F.& Motulsky, A.G. (1986). Human Genetics, 2nd edn. Springer-Verlag, Berlin.

Why should I consider having an amniocentesis?

An amniocentesis will be offered to you if you are at an increased risk of having a baby with a genetic condition. This could be due to a number of reasons including:

- You received an increased risk result on a first or second trimester screening test (see pages 15-17 and 24-26) or non-invasive prenatal testing (NIPT) (see pages 13-15).
- You are over 35 years old. The risk of having a baby with a chromosome condition increases with the mother's age. For further details see "chromosome condition" in the Glossary section of this booklet.
- You or your partner have a family health history of a chromosome condition or other type of genetic condition caused by a gene variation. When the amniocentesis is done to check for a gene variation, the laboratory will need to establish a gene test which can then be used during the amniocentesis. This type of testing is very specialised and is usually organised by a genetic counsellor.

When is an amniocentesis done?

An amniocentesis is usually done between 15 and 19 weeks of pregnancy.

When will the result of amniocentesis be available?

It will take two to three weeks to receive the result of an amniocentesis as it is a lengthy process. After a sample of amniotic fluid has been collected from you, it is sent to a specialised laboratory where cells from the amniotic fluid will be grown in a sterile environment. If the test is looking at the chromosomes, the laboratory may issue an initial result within two to three days. This will provide a result for the common chromosome problems only. The final result will still take two to three weeks.

What will the result of amniocentesis tell me?

It is important to discuss with your doctor or genetic counsellor what conditions the test is able to identify. Depending on the reason why you are having the test, specific conditions can be focused on in the laboratory e.g. if you are an older mother, the main conditions that would be of concern are chromosome conditions.

For other women, they may have previously had a child with a specific genetic condition caused by a gene variation or a specific chromosome change. In this case, the laboratory would analyse that particular gene or look carefully at the chromosome in question in the developing baby.

Although doctors and scientists are now able to diagnose a large number of conditions during pregnancy, it is not possible to test for all of the problems that could affect your developing baby. Your doctor will discuss with you the range of conditions that can be identified using the amniocentesis test and what each of these might mean for you and your baby.

It is important to remember that a normal test result cannot exclude every possible problem with your baby.

How reliable is an amniocentesis?

An amniocentesis is very accurate at identifying the conditions your baby is being tested for.

Very occasionally, more than one amniocentesis might be needed to make a diagnosis. This could happen if the doctor cannot get enough fluid on the first attempt or in rare cases, if the laboratory is unable to get a definite answer. In this situation, a second amniocentesis might have to be done.

What if the result of amniocentesis shows my baby has a problem?

If an amniocentesis result shows that your baby has a problem, you and your partner will be given as much information as possible about the condition. This includes the implications it might have for your baby's future health and development. You will be given time to make an informed choice about whether or not you wish to continue your pregnancy.

If you and your partner decide to continue the pregnancy, you will be offered support and information before, during and after the birth of your baby.

If you decide to terminate your pregnancy, the procedure requires hospitalisation. The method of termination will vary depending on the policy of the particular hospital or clinic. It might involve an induction of labour and birth. All aspects should be fully discussed with your doctor or genetic counsellor. You will be offered support and information before, during and after your hospital stay.

Please see the services support and information section on pages 35-36 of this booklet for more details.

Can an amniocentesis harm me or my baby?

The increase in risk of miscarriage due to having an amniocentesis is less than 1% (1 in 100 pregnancies tested with amniocentesis). This risk is in addition to your "background risk" of miscarriage that all women have in pregnancy due to natural causes. It is important to discuss your background risk of miscarriage with your doctor, midwife or genetic counsellor.

The specific risk figure for a miscarriage depends on the experience of the doctor doing the test and the difficulty he or she has in getting the sample of fluid. It is therefore important that an amniocentesis is only carried out by a doctor experienced in this technique (see the Services: Prenatal testing list on pages 37).

Some women experience cramping and occasionally, vaginal bleeding or leaking of amniotic fluid after an amniocentesis. Talk to your doctor about what you can expect after the testing and what symptoms to look out for.

Second trimester ultrasound

18-20 weeks of pregnancy

- A good way of checking your baby's development
- A safe way of following up on any concerning results that were obtained from an earlier prenatal test
- Does not pose any health risk to you or your baby
- Second trimester ultrasounds, like all other prenatal tests, are optional

What is a second trimester ultrasound?

A second trimester ultrasound will use high frequency sound waves to produce images of your developing baby in your uterus (womb).

The sonographer (a specially trained ultrasound technician) or doctor might spend some time getting the best possible views of the developing baby.

The equipment used to do this type of detailed scan is specialised. It is important that an experienced, well-trained operator with modern equipment performs the scan and interprets the ultrasound images. High quality ultrasound is available from Fetal Medicine Units in public hospitals and from a number of private practitioners. For details of services in your area, see the Services: Prenatal testing list on pages 37.

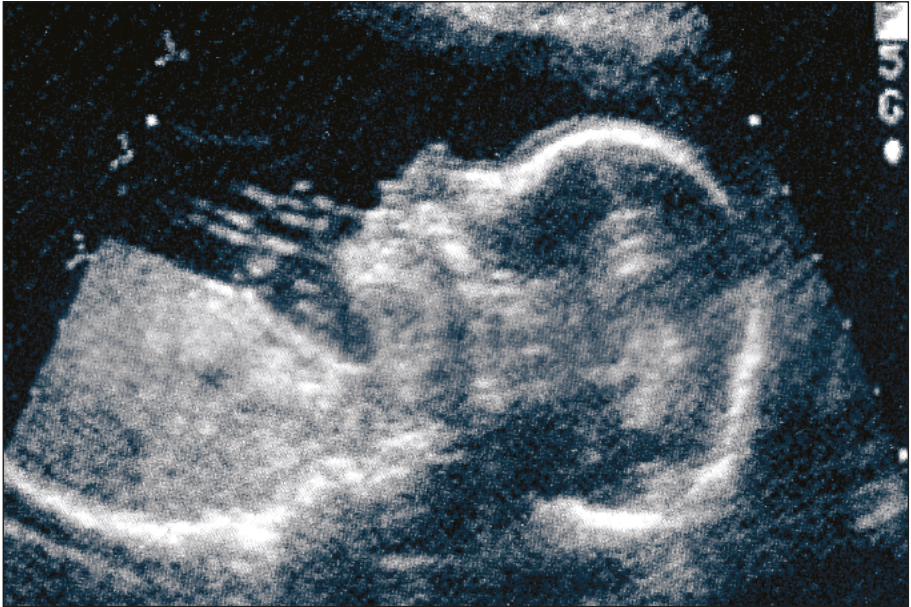
During the ultrasound, the doctor or sonographer will use a water based jelly-like substance and move the transducer (that acts like a microphone) over your abdomen.

Sound waves will pass from the transducer into the uterus and through the fluid surrounding your baby (called amniotic fluid). The sound waves bounce harmlessly off your baby, creating echoes.

A computer will change these echoes into a picture so that the doctor or sonographer can examine your baby's physical development. Your baby won't be able to hear the sound waves as they are very low.

For second trimester ultrasound to be most successful, a full bladder is sometimes needed. The uterus and the ovaries are often hidden behind the bowel, making it difficult to clearly see inside the uterus. However, when the bladder is full, the bowel is pushed out of the way. It is recommended that you have several glasses of non-fizzy fluid one hour before the ultrasound test.

ULTRASOUND SCAN AT 19 WEEKS



Why should I consider having a second trimester ultrasound?

It is common for most pregnant women to be offered a second trimester ultrasound. The main reason this is done is to check on the progress of the baby's growth. Other reasons might be to:

- Check the baby's progress if a first trimester ultrasound raised concern
- Check progress when the baby is at an increased risk of a condition (e.g. because of a family history) that can be diagnosed by ultrasound
- Check the development of the baby's spine if the baby is considered to have a high risk of a neural tube defect such as spina bifida following the second trimester screening test.

When is a second trimester ultrasound done?

A second trimester ultrasound to check the baby's physical development is best done between 18 and 20 weeks of pregnancy.

When will the result of a second trimester ultrasound be available?

In some cases, information about your baby's ultrasound scan will be made available immediately. However, if the person doing the scan suspects that your baby has a problem, he or she might not be able to discuss the findings until they have been verified by another specialist. The result might be sent to the referring doctor who can discuss the result in this situation. Alternatively, you may be referred to a specialty centre for a second opinion and more detailed assessment.

It might be useful to discuss how the result will be given, to whom and when, with the sonographer or doctor before the ultrasound.

What will the result of second trimester ultrasound tell me?

As the second trimester ultrasound will be done at a later stage of pregnancy, your baby's major organs and body structures will be visible. Ultrasound can detect any change to the growth, development, structure and position of the baby's organs. It cannot necessarily detect how well the organs are functioning.

How reliable is second trimester ultrasound?

A second trimester ultrasound cannot check for every physical or intellectual problem that could possibly affect your baby.

What if the result of a second trimester ultrasound shows my baby has a problem?

Sometimes, there may be a subtle indication that the baby *may* have a chromosomal problem. These are sometimes called "soft markers". They are seen in up to 15% of pregnancies but more commonly in babies with a chromosomal condition. Depending on how advanced the pregnancy is, the doctor might discuss the option of having an amniocentesis (see amniocentesis on pages 27-30).

In a small number of cases a second trimester ultrasound scan might show that the baby *has* a genetic condition or some other health or developmental problem. Depending on how advanced the pregnancy is, the doctor might also discuss the option of having a diagnostic prenatal test such as an amniocentesis to give you more information about your baby (see amniocentesis on pages 27-30).

You will be given as much information as possible about the condition that might have been identified. This includes the implications it might have for the future health and development of your baby. You and your partner will be given time to make an informed choice about whether or not you wish to continue your pregnancy.

If you decide to continue your pregnancy following the diagnosis of a problem with your baby, you will be offered support and information before, during and after the birth of your baby.

If you decide to terminate your pregnancy, the procedure requires hospitalisation. The method of termination will vary depending on the policy of the particular hospital or clinic. It might involve an induction of labour and birth. All aspects should be fully discussed with your doctor, midwife or genetic counsellor.. You will be offered support and information before, during and after your hospital stay.

Please see the Services: Support and Information section on pages 37-40 of this booklet for more details.

Can a second trimester ultrasound harm me or my baby?

No evidence has been found that shows an ultrasound can harm you or your baby.

Pre-pregnancy option Preimplantation genetic diagnosis (PGD)

- Involves testing for certain genetic conditions in an embryo created using in-vitro fertilisation (IVF)
- Only those embryos that do not have the specific genetic condition that was tested for will be transferred into the woman's uterus
- Currently only offered in the private health setting

What is preimplantation genetic diagnosis (PGD)?

Preimplantation genetic diagnosis (PGD) is a very specialised technique that can help couples who are at risk of passing on a genetic condition to their children avoid doing so. PGD involves testing an embryo that has been created using in-vitro fertilisation (IVF), prior to transferring it to the mother's uterus and allowing it to develop.

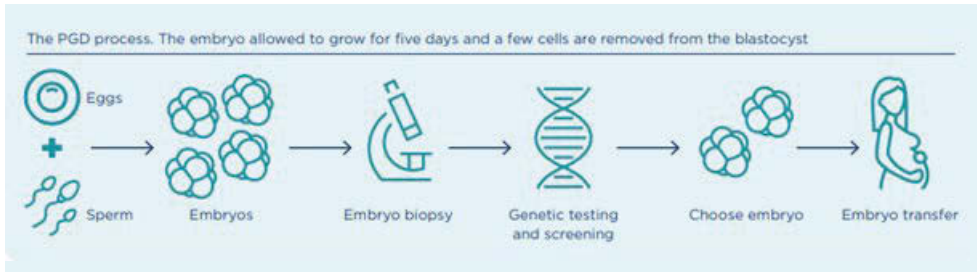
How and when is preimplantation genetic diagnosis (PGD) done?

Hormones are used to stimulate a woman's ovaries and enable the collection of a number of eggs (oocytes). The eggs are then fertilised with sperm in the laboratory. Those eggs that are successfully fertilised divide and multiply to form a developing embryo. After several days, the embryo has formed into a ball containing many more cells.

Once the embryo reaches this stage, one or two cells are removed and tested for the specific genetic condition in question.

Only those embryos that are found not to be affected by the specific genetic condition will be transplanted into the mother's uterus. Usually, no more than one or two embryos will be transferred to the uterus at any one time to avoid the possibility of multiple births. In some IVF centres, unaffected embryos that are not used can be frozen for transfer at a later stage.

What information does PGD provide?



Success rates for having a child following IVF and PGD varies from centre to centre but tend to follow standard IVF success rates. The addition of a PGD step to IVF, however, might limit the number of embryos available for transfer. For this reason, a pregnancy and an unaffected child cannot be guaranteed using this technique. It is important for an IVF centre to have a high pregnancy rate to be able to offer reliable PGD with a good chance of achieving pregnancy.

Like any IVF procedure, stress and often disappointment can accompany PGD. Couples will need to consider the financial and emotional burden of using IVF and PGD versus the burdens associated with terminating an affected child conceived naturally.

For couples with a moral or religious objection to pregnancy termination and who also have a risk of having a child with a genetic condition, this technique might provide the opportunity to have an unaffected child. It can also help some couples avoid having repeated miscarriages when one partner carries a particular chromosome condition.

In Australia, PGD is only offered through private clinics. IVF centres offering this service can be found on page 39.

SERVICES

Prenatal testing services within NSW

CAMPERDOWN

Royal Prince Alfred Hospital
RPA Women & Babies
Fetal Medicine Unit
Ph: 02 9515 6042

KOGARAH

St George Hospital
Women's & Children's Health
Ph: 02 9113 1495

LIVERPOOL

Liverpool Hospital
Fetal Medicine Unit
Ph: 02 8738 5631

NEWCASTLE

John Hunter Hospital
Maternal Fetal Medicine Unit
Ph: 02 4921 4694

PENRITH

Nepean Hospital
Perinatal Ultrasound
Ph: 02 4734 2578

RANDWICK

Sydney Children's Hospital
Centre for Clinical Genetics
Ph: 02 9382 5608

Royal Hospital for Women
Maternal Fetal Medicine
Ph: 02 9382 6098

ST LEONARDS

Royal North Shore Hospital
Fetal Medicine Unit
Ph: 02 9463 2370

WESTMEAD

Westmead Hospital
Fetal Medicine Unit
Ph: 02 8890 6802

Genetics services within NSW

CAMPERDOWN

Royal Prince Alfred Hospital
Molecular and Clinical Genetics Unit
Ph: 02 9515 5080 Fax: 02 9515 5490

KOGARAH

Clinical Genetics Service
St George Hospital
Ph: 02 9113 3635 Fax: 02 9113 3694

DARLINGHURST

St Vincent's Hospital
St Vincent's Clinical Genomics
Ph: 02 8382 4899 Fax: 02 8382 4895

LIVERPOOL

Liverpool Hospital
Dept of Clinical Genetics
Ph: 02 8738 4665 Fax: 02 8738 4650

NEWCASTLE

Hunter Genetics
Regional Medical Genetics Unit
Ph: 02 4985 3100 Fax: 02 4985 3105

PENRITH

Nepean Hospital
Clinical Genetics Department
Ph: 02 4734 3362 Fax: 02 4734 4472

RANDWICK

Sydney Children's Hospital
Centre for Clinical Genetics
Ph: 02 9382 5608 Fax: 02 9382 5683

ST LEONARDS

Royal North Shore Hospital
Dept of Clinical Genetics
Ph: 02 9463 1727 Fax: 02 9463 1057

WESTMEAD

Children's Hospital at Westmead
Dept of Clinical Genetics
Ph: 02 9845 3273 Fax: 02 9845 3204

WESTMEAD HOSPITAL

Department of Genetic Medicine
Ph: 02 8890 9780 Fax: 8890 9789

NSW outreach genetic services

BATHURST

Bathurst Health Service
Community Health Centre
Howick Street, Bathurst NSW 2795
Ph: 02 6330 5677 Fax: 02 6331 0474

COFFS HARBOUR

Child Adolescent & Family Service
Locked Mail Bag 812
Coffs Harbour NSW 2450
Ph: 02 6656 7200 Fax: 02 6656 7203

DUBBO

Dubbo Community Health Centre
2 Palmer Street
DUBBO NSW 2830
Ph: 02 5853 2500 Fax: 02 5853 2580

FORSTER

Forster Community Health Centre
Breese Pde, Forster NSW 2428
Ph: 02 6539 6300 Fax: 02 6554 8874

GOSFORD

Contact Royal North Shore Hospital
Ph: 02 9463 1727 Fax: 02 9463 1057

GOULBURN

Child Infant and Family Tertiary Service
Locked Bag 2015, Goulburn NSW 2580
Ph: 02 4825 4050 Fax: 02 4825 4058

KEMPSEY

C/- Port Macquarie Community Health Ctr
Ph: 02 6589 2328 Fax: 02 6589 2160

KINGSCLIFF

Kingscliff Community Health
Turnock Street, Kingscliff NSW 2487
Ph: 02 6674 9500 Fax: 02 6674 9599

PORT MACQUARIE

Port Macquarie Health Campus Morton
Street
Port Macquarie NSW 2444
Ph: 02 6589 2328 Fax: 02 6589 2160

TAMWORTH

Tamworth Community Health Centre
Locked Bag 9783
NEMSC NSW 2348
Ph: 02 6767 8151 Fax: 02 6766 3967

TAREE

Taree Community Health
PO Box 35, Taree NSW 2430
Ph: 02 6592 9315 Fax: 02 6592 9607

WAGGA WAGGA

Wagga Wagga Base Hospital
PO Box 159, Wagga Wagga NSW 2650
Ph: 02 5943 4043 Fax: 02 6933 9262

WOLLONGONG

Northern Illawarra Maternity and
Women's Health
Wollongong Hospital
Crown Street, Wollongong NSW 2500
Ph: 02 4253 4267 Fax: 02 4253 4257

Interstate genetics services

AUSTRALIAN CAPITAL TERRITORY

ACT Genetics Services
Canberra Health Services
PO Box 11, Woden ACT 2606
Ph: 02 5124 7630 Fax: 02 5124 3021

NORTHERN TERRITORY

Royal Darwin Hospital and
Alice Springs Hospital
Ph: 08 8944 8731 Fax: 08 8922 8463

QUEENSLAND

Genetic Health Qld
Royal Brisbane and Women's Hospital
Herston QLD 4029
Ph: 07 3646 1686 Fax 07 3646 1987
Email: GHQ@health.qld.gov.au
Web: <https://metronorth.health.qld.gov.au/rbwh/genetic-health-queensland>

SOUTH AUSTRALIA

South Australian Clinical Genetics Services
72 King William Road,
North Adelaide SA 5006
Ph: 08 8161 7375 Fax: 08 8161 7754

TASMANIA

Tasmanian Clinical Genetics Service
C/- Royal Hobart Hospital
GPO Box 1061L, Hobart TAS 7001
Ph: 03 6166 8296 Fax: 03 6173 0328

VICTORIA

Victorian Clinical Genetics Services
Royal Children's Hospital
Parkville VIC 3052
Ph: 03 8341 6201 Fax: 03 8341 6390

WESTERN AUSTRALIA

Genetic Services of WA
King Edward Memorial Hospital for Women
374 Bagot Road, Subiaco WA 6008
Ph: 08 6458 1525 Fax: 08 6458 1678

Pre-implantation Genetic Diagnosis (PGD) Providers in NSW

IVF AUSTRALIA

Level 1, 33 York St, Sydney NSW 2000
Freecall 1800 111 483
Ph: 02 8346 6800 Fax: 02 9231 1010
Web: www.ivf.com.au

GENEA

321 Kent St,
Sydney, NSW 2000
Ph: 1300 361 795
Web: www.genea.com.au

MONASH IVF

Level 2, 1 Fennell St,
Parramatta, NSW 2151
Ph: 02 9890 9022
Web: www.monashivf.com

CITY FERTILITY CENTRE

Level 1, Suite 101, 1 Moore St,
Liverpool, NSW 2170
Ph: 1300 354 354
Web: www.cityfertility.com.au

ROYAL HOSPITAL FOR WOMEN

Fertility and Research Centre
Ph: 02 9382 6666
Web: www.fertilityandresearchcentre.com.au

HUNTER IVF

Suite 4, The Heights Private Medical Centre
Newcastle Private Hospital
2 Lookout Rd, New Lambton Hts NSW 2305
Ph: 02 4957 8515 Fax: 02 4952 3859
Web: <https://www.ivf.com.au/clinics/hunter-ivf>

WESTMEAD FERTILITY CENTRE

Level 2 (Ground Floor), G Block
Westmead Hospital
Westmead, NSW 2145 Ph: 02 8890 7484
Web: <https://www.westmeadfertilitycentre.com.au/>

Support and Information

Please see the [Support page](#) on the Centre for Genetics Education website for key information at

<https://www.genetics.edu.au/SitePages/Support-groups.aspx>

The Centre for Genetics Education

produces a number of resources for health professionals and their patients. These include two booklets which are only available online:

- [“Diagnosis of abnormality in an unborn baby – The Impact, Options and Afterwards”](#)
- [“When your unborn baby has a problem – How to manage the weeks ahead”](#)

DOWN SYNDROME ASSOCIATION

provides a parent support network, information and resources, and peer support for families and people with Down syndrome. Resources are available in many languages.

Down Syndrome New South Wales
Level 6, 410 Church St
North Parramatta NSW 2151
Ph: 02 9841 4444
Email: admin@dsansw.org.au
Website: <https://www.downsyndrome.org.au/nsw/>

GENETIC ALLIANCE AUSTRALIA (GAA)

provides information and support to families and individuals who are affected by a rare genetic condition. GAA has a client database representing over 500 genetic conditions.

Genetic Alliance Australia
C/- Garvan Institute of Medical Research
Level 6, 384 Victoria Street,
Darlinghurst NSW 2010
Ph: +61 2 9295 8359
Email: info@geneticalliance.org.au
Website: www.geneticalliance.org.au

RARE VOICES AUSTRALIA

National peak body for Australians living with a rare disease. RVA advocates for policy and key systems to help people with a rare condition.

Website: <https://rarevoices.org.au/>

SANDS Australia provides miscarriage, stillbirth and newborn death support. They also offer support to individuals experiencing a prenatal diagnosis of an abnormality or condition.

Sands
Suite 4/670 Canterbury Road
Surrey Hills, Victoria, 3127
Ph: 1300 998 698
Website: www.sands.org.au Email: support@rednose.org.au

THE SPINA BIFIDA GROUP provides support, information and a peer support network for families and people with spina bifida. There are branches of the association in all states of Australia.

Northcott
1 Fennell Street,
North Parramatta NSW 2151
Ph: 1800 818 286
Email: northcott@northcott.com.au
Website: www.northcott.com.au

THE NATIONAL ASSOCIATION FOR LOSS AND GRIEF (NALAG)

promotes loss and grief support groups throughout Australia.

National Association For Loss
And Grief (NALAG)
PO Box 379
Dubbo NSW 2830
Ph: 02 6882 9222
Fax: 02 6884 9100
Email: info@nalag.org.au
Website: www.nalag.org.au

GLOSSARY

Terms that are underlined appear as separate entries in the Glossary

AMNIOTIC FLUID

Transparent, almost colourless fluid contained within the sac surrounding the developing baby. Cells from the baby are found in this fluid and can be grown in a laboratory and used for testing.

ANENCEPHALY

See [neural tube defects](#).

CHORION

The chorion forms part of the [placenta](#) and develops from the fertilised egg.

CHROMOSOMES

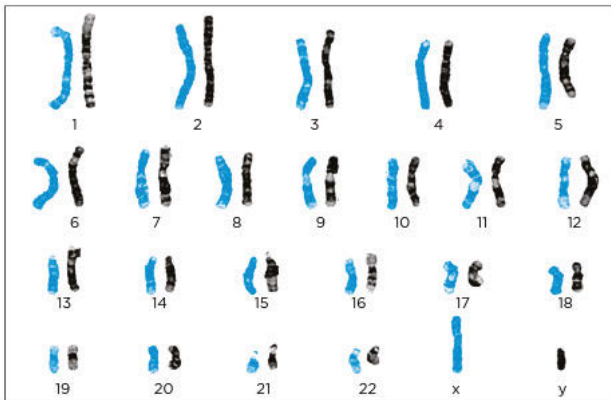
Chromosomes are tiny structures present in every cell of the body. Each chromosome contains thousands of [genes](#) that are essential to an individual's health, growth and development. In each normal human cell, except the egg and the sperm, there are 46 chromosomes, each of which is represented twice giving 23 different chromosome pairs. One of the chromosomes in each pair comes from the mother and the other from the father.

The chromosomes vary in size and scientists have given them a number based on that size from 1 to 22. These are called autosomes. The remaining 2 chromosomes are the sex chromosomes, called X and Y. Females have two copies of the X chromosome (XX) and males have one X and one Y chromosome (XY).

In the normal reproductive cycle, the mother's egg - containing 23 chromosomes is fertilised by the father's sperm - also containing 23 chromosomes. This union results in an embryo containing 46 chromosomes which will develop into the baby.

Figure 1 (see next page) is called a karyotype. It is a picture of an individual's chromosomes. The chromosomes are stained with special dyes in the laboratory to produce the distinct banding patterns. Arranging the chromosomes by size makes systematic study possible. Here, there are 22 pairs of autosomes (numbered chromosomes) and one pair of sex chromosomes. This is the karyotype of a male because there is an X and a Y chromosome.

FIGURE 1: A NORMAL MALE KARYOTYPE (46,XY)



DIAGNOSTIC TEST

A diagnostic test looks for a specific genetic condition that your baby might be at risk of. These types of tests can assess your baby for a [chromosome condition](#) or a condition caused by a variation in a single gene. A diagnostic test does not check every possible physical or intellectual problem that could affect your baby.

Prenatal diagnostic tests include:

- Ultrasound
- Chorionic villus sampling (CVS)
- Amniocentesis

DOWN SYNDROME

Down syndrome occurs when there is an extra copy of chromosome number 21, this is why it is also known as trisomy 21. There are certain physical characteristics associated with Down syndrome and varying degrees of intellectual disability.

EMBRYO

An egg that has been fertilised by a sperm.

GENES

Genes provide the information for our growth, development and health. Everyone has variations in their genetic information, which is why we are all unique. Most gene variations cause no problem.

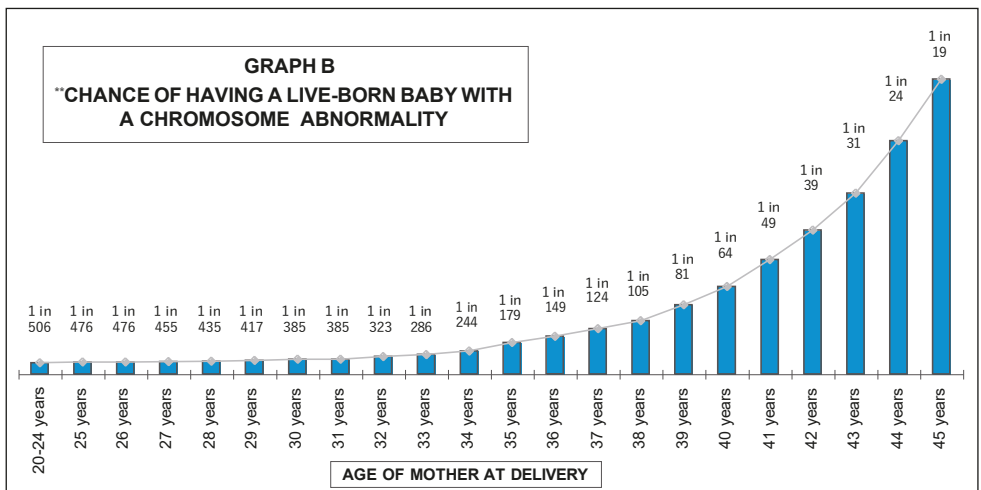
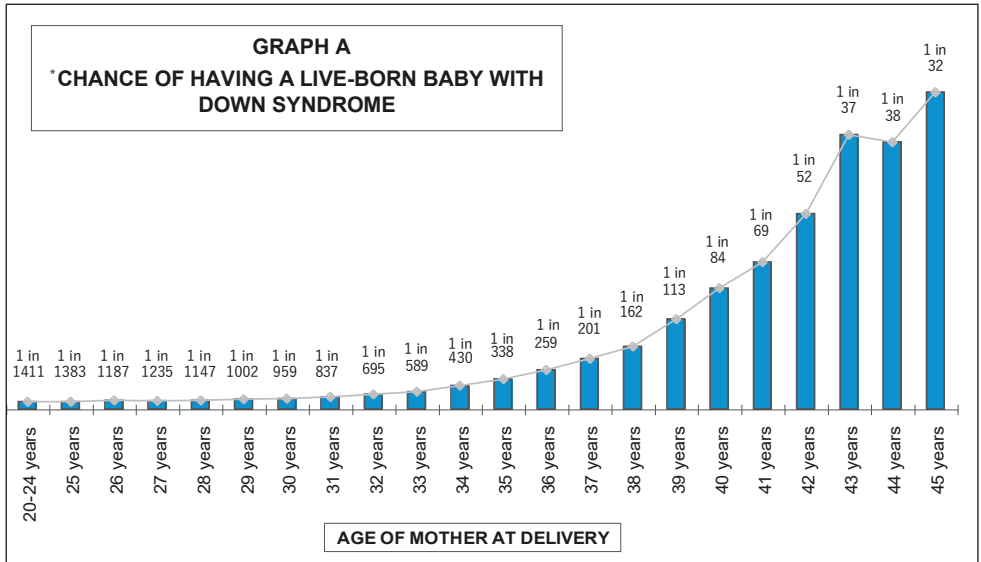
- *Faulty Genes:* Some variations in the genetic information make the gene faulty and are called mutations. Faulty genes do not work as they should in the body. A faulty gene can be inherited from one or both parents or can occur in the formation of the egg or sperm or at conception. The pattern of inheritance of the faulty gene can sometimes be traced through the family health history.

GENETIC CONDITIONS

Genetic conditions include many of the growth, developmental and health problems seen at birth. Others may not be noticed until childhood, adolescence or adulthood.

In addition to chromosome conditions, there are over 6,000 other known genetic conditions that are due to faulty genes. A faulty gene can be inherited in a family and passed from one generation to another.

As there are two copies of every gene (one on each of the chromosomes in a pair), one copy may be faulty and not cause a problem as the other copy is still working as it should. We are all carriers of a small number of faulty genes.



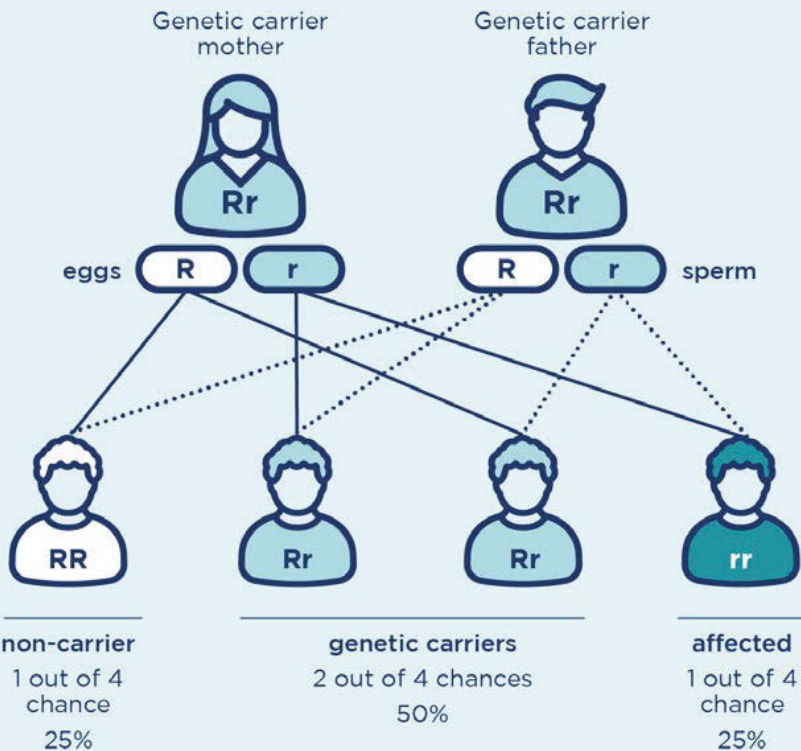
The three most common patterns of genetic inheritance are described below:

(A) Autosomal recessive inheritance

As shown above when carriers of the same faulty gene have a baby, there is a 1 in 4 chance (or 25%) that they will have a child who gets both copies of the faulty gene from the parents. In this case, the expression of the faulty gene is not masked by the correct gene copy and the baby will be affected at some time in life. The 1 in 4 chance applies to every pregnancy.

DIAGRAM OF AUTOSOMAL RECESSIVE INHERITANCE

Autosomal recessive inheritance when both parents are unaffected genetic carriers for the condition. The non-working copy of the gene containing a recessive variant is represented by 'r'; the working copy of the gene by 'R'.



(B) Autosomal dominant inheritance

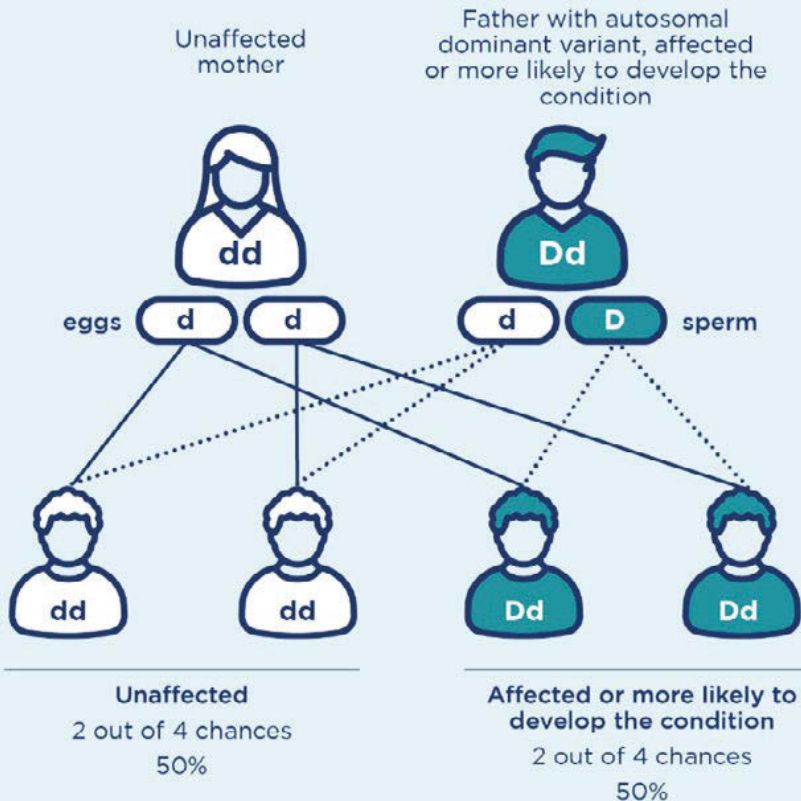
Some faulty genes override the effect of the working gene copy on the other chromosome. The result may be a genetic condition that is seen at birth or later in life.

Where one of the parents is affected by a condition due to a faulty gene, they can pass on to the baby either the correct or the faulty gene copy.

As shown below there is a 1 in 2 (or 50%) chance in every pregnancy that the faulty gene will be passed on and the baby will be affected by the condition at some time in life. These risks apply to every pregnancy.

DIAGRAM OF AUTOSOMAL DOMINANT INHERITANCE

Autosomal dominant inheritance - when one parent has the autosomal dominant non-working gene copy. The non-working gene copy with an autosomal dominant variant is shown as 'D'; the working copy of the gene by 'd'.

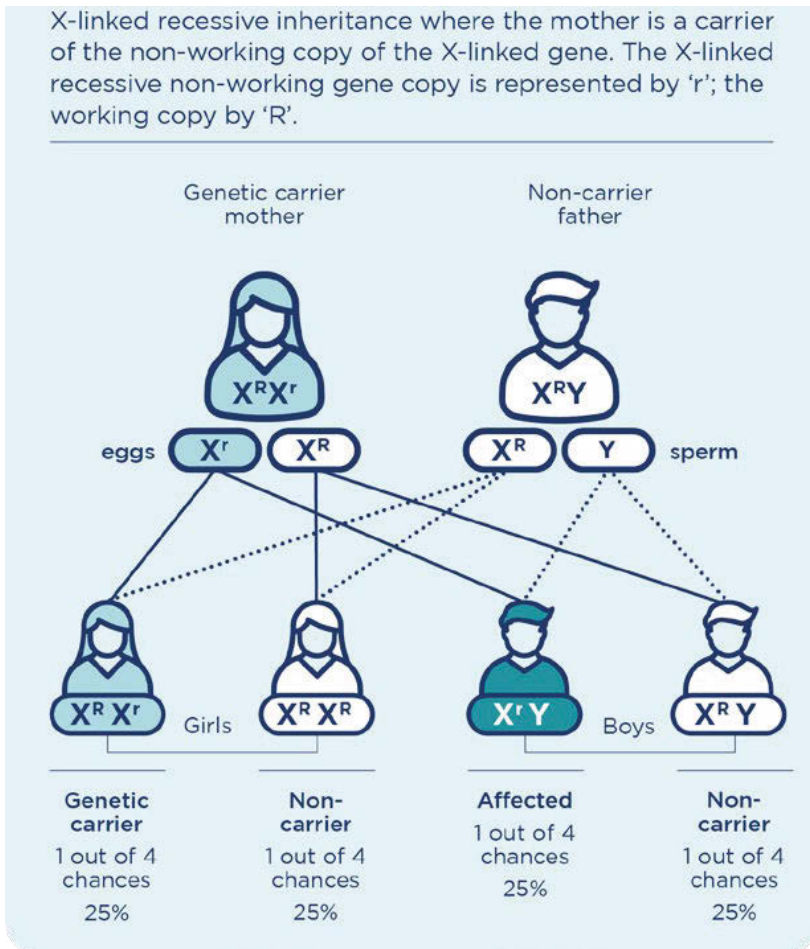


(C) X-linked inheritance:

Where genetic conditions are due to a faulty gene carried on the X chromosome (X-linked), the condition affects males and females differently. There have been very few genes located on the Y chromosome. Since males are XY, they have only one X chromosome copy. A faulty gene carried on the X chromosome would therefore be expressed, causing a genetic condition, as they do not have a correct gene copy to override the faulty gene. Females have two X chromosome copies and therefore can be carriers of the faulty gene but are not usually affected.

Where the mother is a carrier of the faulty gene and the father is unaffected, there is a 1 in 2 chance (or 50%) that their female children will also be carriers but will be unaffected. However, there is a 1 in 2 chance (or 50%) that their male children will inherit the faulty gene and be affected. These risks apply to every pregnancy.

DIAGRAM OF X-LINKED INHERITANCE



IN-VITRO FERTILISATION (IVF)

A medical procedure that involves removing eggs from a woman's ovaries and fertilising them with a man's sperm in a laboratory. The resulting [embryo\(s\)](#) are then transferred into the woman's uterus through the cervix with the aim of achieving a pregnancy.

NEURAL TUBE DEFECTS

The most common types of neural tube defects are [anencephaly](#) and [spina bifida](#).

- Anencephaly is the most serious as the baby's brain does not develop properly. Babies with anencephaly do not survive.
- Spina bifida refers to an opening on the baby's spine which exposes the spinal cord and can cause paralysis and other problems.

The majority of these neural tube defects may be prevented by the supplementation of the mother's diet with 0.5mg of the vitamin folate or folic acid for at least one month prior to pregnancy and continuing for the first three months of the pregnancy.

Women at an increased risk for having a baby with a neural tube defect as described below will need to take a higher dose of folate. This should be discussed with your doctor.

In Australia, neural tube defects affect about 1 in 500 babies. The risk for having a baby with a neural tube defect may be increased if:

- There is a close family health history of one or more people affected by a neural tube defect. This can be either on the mother's or father's side of the family.
- A woman has insulin-dependent diabetes
- A woman is taking specific medicine to control seizures (epilepsy).

Genetic counselling is recommended for these women.

PLACENTA

The placenta, sometimes called afterbirth, is the organ on the wall of the uterus to which the developing baby is attached by the umbilical cord. The placenta is involved in basic nourishment of the developing baby and in hormone production.

SCREENING TEST

A screening test cannot tell if your baby definitely has a [genetic condition](#) but they might indicate that further testing needs to be considered (See [diagnostic test](#)).

Prenatal screening tests include:

- Ultrasound
- NIPT (non-invasive prenatal testing)
- First trimester screening - nuchal translucency
- Second trimester screening - maternal serum testing

SPINA BIFIDA

See [neural tube defects](#).

TERMINATION OF PREGNANCY

A medical procedure to end a pregnancy prior to its natural completion. Also known as an abortion.

TRISOMY 21

See [Down syndrome](#)

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