Prenatal testing and genetic counselling

Every couple wants to have a healthy baby. However, there are some couples whose baby may have (or will develop) a serious physical and/or intellectual condition. There are a number of different tests available to assess the health and development of a baby before birth. Each has advantages, disadvantages and limitations. The decision to undertake testing during a pregnancy is a very personal one. It's also a decision best made on all the available information.

What are prenatal tests?

Prenatal tests are tests done while you are pregnant to assess the health and development of your baby. There are two main types of prenatal tests:

Screening tests
A screening test can indicate that there's a need for further testing (called diagnostic testing), but it can't tell if your baby definitely has a particular condition. You can choose whether or not you have a prenatal screening test.

Screening tests may be performed from 11 weeks of pregnancy and may include:

- ultrasound examination of developing baby and blood tests for you
- a nuchal (pronounced new-cal) translucency ultrasound which is sometimes combined with a blood test for the mother. This screening test is performed in the first trimester
- a maternal serum test which tests the mother's blood in the second trimester

Prenatal diagnostic tests
Diagnostic tests look for more specific conditions that your baby might be at risk. They are generally only performed if a screening test identifies an increased risk of a baby having a particular condition (although women over 35 can choose to have a diagnostic test). These types of tests can assess your baby for a chromosome condition or a condition caused by a variation in a single gene (these are called genetic conditions). A diagnostic test does not check every possible physical or intellectual problem that could affect your baby.

Diagnostic tests include chorionic villus sampling (CVS) and amniocentesis (pronounced am-nee-o-cen-tee-sis)

Each prenatal test is done at a certain time during the pregnancy. The tests and their timings are shown in the table *Prenatal Screening and Diagnostic Testing* on pages 118-119.

What information does a prenatal test provide?

A prenatal test may be done to check if your baby is developing in the usual way. It can also be performed to see if your baby is at risk of (or is affected by) a specific physical and/or intellectual condition.

Some of the conditions that prenatal tests are able to detect include **genetic conditions.**

Genetic conditions include many of the physical and/or intellectual conditions that are found at birth, in childhood, adolescence or adulthood. A genetic condition can occur for a number of reasons. Prenatal tests identify the sorts of conditions that are 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 of genetic information or the way it's arranged 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 such changes is unknown. The most commonly known chromosome condition is Down Syndrome.

Other genetic conditions, which are caused by a variation that makes a single gene faulty, are known to affect babies in some families. A couple may already have a child with one of these conditions, or one of the parents may have the condition themselves. In these situations, a prenatal test can be used to specifically identify if the baby has the faulty gene that causes the genetic condition.

Prenatal Testing: Special tests for your baby during pregnancy is a detailed booklet produced by the Centre for Genetics Education. To obtain a copy, contact Tel: (02) 9462 9599 or visit http://www.genetics.edu.au/ Information about these tests is also available in other languages from http://www.genetics.edu.au/Information/multilingual-resources

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

A booklet called *Your choice: screening and diagnostic tests in pregnancy* is available online. It talks about the different tests that are available, explains the conditions that can be detected and gives you some things to think about to help you decide about testing during your pregnancy. You can find it at http://www.mcri.edu.au/Downloads/PrenatalTestingDecisionAid.pdf

Why would I consider having a prenatal test?

Before you make any decision about having a prenatal test, you should get as much information as possible so that you feel confident you're making the right choice for you. You need to be able to discuss your concerns and thoughts about prenatal testing with professionals in a safe and understanding environment. Getting help and advice at this time can help you make informed decisions about the future of your pregnancy.

It is important to discuss:

- 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.

What if a test shows my baby may have a problem?

If the result of a prenatal test shows that your baby is not developing normally or could develop a problem after birth, you and your partner will be given as much information as possible about the condition. Genetic counselling will give you the opportunity to discuss:

- what the results mean for your baby and your family
- the options for further testing and what it involves
- your feelings about people with physical and/or intellectual disabilities
- your feelings about termination of pregnancy.

If you want to know more about prenatal testing and genetic counselling, contact the Centre for Genetics Education on (02) 9462 9599 or visit www.genetics.edu.au

What is genetic counselling?

Genetic counselling is available at most large hospitals and many local Community Health Centres. It is provided by a team of health professionals who work together to give you current information and counselling regarding genetic problems in the growth, development and health of your baby. It can help you to understand and make informed choices about special prenatal testing during pregnancy or adjust to the diagnosis of a genetic condition in your baby.

Who should consider having genetic counselling early in pregnancy?

When you are pregnant it is important to discuss with your partner any health concerns you have and whether you or other members of your family have medical problems which may run in the family.

There are a number of situations in which genetic counselling may be helpful. These include:

- you or your partner have a close relative or a child with a physical and/or intellectual condition
- you or your partner has a serious condition that may be passed on to a baby
- you and your partner are carriers of the same faulty gene
- you are in your older than 35 (not necessarily your first pregnancy) as there is an increased risk for having a baby with chromosome condition
- you have been exposed to a chemical or other environmental agent during this pregnancy
- the results of a screening test such as ultrasound or first and second trimester screening have suggested that your baby is at increased risk for a particular genetic condition
- you and your partner are related e.g. first cousins
- you and your partner are both from an ethnic or cultural background which is known to carry certain genes for a common condition in that population.

Knowing about your family health history is important

Health-related information about you and your partner's parents, brothers and sisters is valuable information that can identify some health conditions you might be at an increased risk of developing or passing on to your children. In some cases, you might be able to take preventative measures to reduce your risk and that of future generations.

There are many health conditions that can be passed on by family members. However, it is important to remember that lifestyle factors (e.g. smoking or an unhealthy diet) can also play a part in the chances of developing a condition.

When discussing your family health history with your relatives, you should think about some well-known conditions such as:

- hearing problems
- learning or developmental problems
- cystic fibrosis – a genetic condition which affects lung and digestive function
- thalassaemia – a genetic condition that affects normal blood processes
- sickle cell disease – a genetic condition that leads to serious blood anaemia
- haemophilia – a genetic condition affecting boys that leads to excessive bleeding
- heart disease
- diabetes
- cholesterol problems
- breast, ovarian or bowel cancer.

It is also important to think about the geographic background of your family and that of your partner. For example, people from Northern Europe and the United Kingdom are more likely to carry a faulty gene that causes cystic fibrosis than thalassaemia which affects more people who come from the Middle East and the Pacific.

More information about collecting your family health history and some of the conditions mentioned above is available by contacting the Centre for Genetics Education on (02) 9462 9599 or visiting www.genetics.edu.au

Prenatal Screening and Diagnostic Testing

Stage of pregnancy	Name of test	Type of test	How is the test done?
8-12 weeks	First trimester ultrasound – dating scan.	Screening and diagnostic.	A small probe is pressed on to the mother's abdomen or inserted into the vagina. This shows a picture of the developing baby.
11-13 weeks	Chorionic villus sampling (CVS).	Diagnostic.	With the help of ultrasound, a small sample of the placenta is taken through the mother's abdomen, using a thin needle or through the cervix using a thin flexible tube.
11.5-13.5 weeks	Nuchal translucency (ultrasound) test with or without testing of the mother's blood.	Screening.	Using an ultrasound, a special measurement (nuchal translucency) is taken of the baby. Also, a sample of the mother's blood may be taken for testing.
15-18 weeks	Second trimester screening test – maternal serum testing.	Screening.	A sample of the mother's blood is taken for testing.
15-19 weeks	Amniocentesis.	Diagnostic.	With the help of ultrasound, a small sample of the amniotic fluid is taken through the mother's abdomen, using a thin needle.
18-20 weeks	Second trimester ultrasound – fetal anomaly scan .	Screening and diagnostic.	An instrument like a microphone is pressed on to the mother's abdomen. This shows a picture of the developing baby.

	What does this test look for?	Are there any risks to the baby or mother?
	This test can check the size of the baby, check if there is more than one baby and see some physical features such as the heartbeat.	This test does not harm the baby or the mother.
	This test can check for a range of physical and/or intellectual conditions that the baby may have. These are known as chromosomal problems. Sometimes more testing may be needed.	There is a small chance (less than 1 in a 100) that this test may cause a miscarriage. The mother may feel some discomfort during the test.
	This test can tell if a baby has an increased risk of certain physical and/or intellectual conditions. About 5 out of a 100 babies tested may have an increased risk result. Most of these babies will **NOT** have a problem. If the nuchal translucency test is done without the blood test: • About 75 out of a 100 babies who have a chromosome condition called Down Syndrome will return an increased risk result. 25 out of a 100 babies with Down Syndrome will be missed by this test. If the nuchal translucency ultrasound is done together with the blood test: • About 80-90 out of a 100 babies who have Down Syndrome will return an increased risk result. 10-20 out of a 100 babies with Down Syndrome will be missed by this test.	This test does not harm the baby or the mother.
	This test can tell if a baby has an increased risk of certain physical and/or intellectual conditions. About 5 out of a 100 babies tested may have an increased risk result. Most of these babies will **NOT** have a problem. About 60 out of a 100 babies who have Down Syndrome will return an increased risk result. 40 out of a 100 babies with Down Syndrome will be missed using this test. If this test is done at the same time as a detailed ultrasound scan, it will also identify about 95 out of a 100 babies who have spinal problems or neural tube defects.	This test does not harm the baby or the mother.
	This test can check for a range of physical and/or intellectual conditions that the baby may have, including chromosome conditions.	There is a small chance (less than 1 out of a 100 babies) that this test may cause a miscarriage. The mother may feel some discomfort during the test.
	This test can check the size of the baby and many physical features such as the heart, brain, spine and kidney development.	This test does not harm the baby or the mother.