Genetics Fact Sheet

PRENATAL TESTING AND SCREENING FOR FETAL ABNORMALITIES

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WHAT IS PRENATAL TESTING AND SCREENING?

Prenatal testing is the use of special tests carried out during pregnancy. There are two kinds of tests.

Diagnostic tests determine if the baby has, or will develop after birth, a particular problem. These problems include problems in growth and development and problems in the way in which the body functions. It is possible to detect conditions that are due to changes in chromosome number and structure as well as changes to the genetic information in particular genes (see Genetics Fact Sheets 1, 12 & 13). Some of the conditions that can be determined during pregnancy would be evident at birth or in the first year of life (birth defects) but others would not affect a person until later in childhood, adolescence or even as an adult.

Screening tests determine if the baby has an increased risk of having a particular problem. They are not diagnostic tests and an increased risk result does not mean the baby will definitely have a problem.

WHY SHOULD PRENATAL DIAGNOSIS BE CONSIDERED?

There are a number of reasons that would suggest the consideration of prenatal diagnostic testing.

These include:

- having a family history of a person with a serious condition
- both parents are "carriers" of the same faulty gene
- where one of the partners in a couple has a serious condition which may be passed on to a baby
- having a previous child affected by a serious problem in growth, development and/or health
- having a family history of a condition that only affects the male members of a family. Women in the family may be "carriers" of the faulty gene for the condition in question (see Genetics Fact Sheets 1 & 2).
- where a woman is having a baby when she is in her mid-thirties or older (not necessarily her first pregnancy)
- exposure to some chemical or other environmental agent
- where the results of screening tests such as ultrasound or first and second trimester screening tests (see later) have determined that the woman is at increased risk for having a baby with a particular genetic condition in this pregnancy.

Prenatal screening tests are available to pregnant woman of all ages, in particular to those women who do not have any of the above indications.

CAN ALL BIRTH DEFECTS BE DETECTED BY PRENATAL DIAGNOSIS?

No. It is not possible to detect or diagnose every possible problem a child might have using prenatal diagnosis.

HOW IS PRENATAL TESTING DONE?

There are an increasing number of tests available to detect birth defects and other genetic conditions or determine if a pregnancy is at increased risk for these problems. Some are screening tests using ultrasound and/or an examination of chemicals in the mother's blood to determine whether the baby is at risk of certain problems. Other tests are diagnostic as they examine cells from the baby to determine if the baby actually has the condition in question.

Where IVF (in vitro fertilisation) is used to conceive a child, testing may be available for certain conditions prior to the embryo being implanted in the mother's uterus. This type of testing is called preimplantation diagnosis (see Genetics Fact Sheet 16).

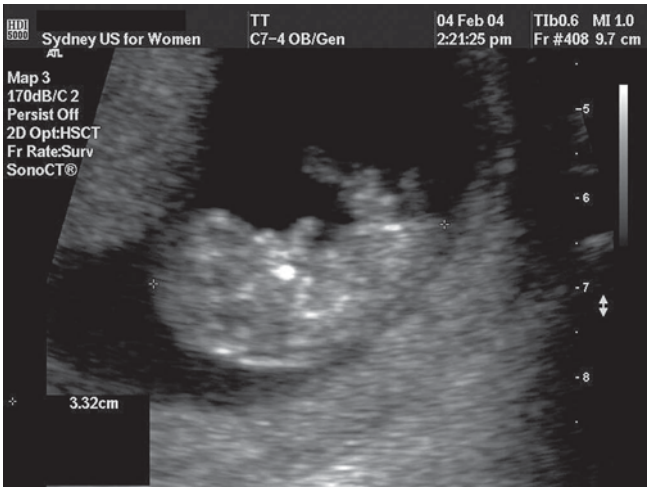

Figure 1: Ultrasound at 10 weeks, showing the whole baby

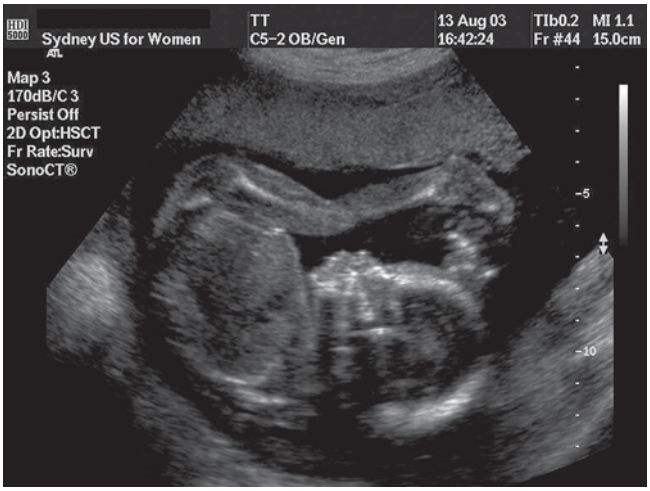

Figure 2: Ultrasound at 18 weeks, showing whole baby

Genetic counselling (see Genetics Fact Sheet 5) can provide the most up to date information about the availability and use of tests prior to and during pregnancy. It can provide an opportunity for an informed decision to be made about the utilisation of prenatal diagnostic testing.

There are a number of different tests and procedures that can be performed during pregnancy. Each has distinct advantages, disadvantages and limitations. Prenatal diagnostic testing, prenatal screening and ultrasound scanning are outlined below in the order in which they are offered.

8-10 weeks and 18 weeks
*ULTRASOUND SCANNING (*See Figures 1 and 2*)*
Can be used as a screening and a diagnostic test

Almost every woman has an ultrasound scan at some time during pregnancy. Sound waves, which do not harm the baby, are passed through the amniotic fluid surrounding the baby to create a picture.

- Ultrasound can be performed at any stage during the pregnancy and is usually done at 8-10 weeks to check the dates of the pregnancy and the number of babies present.
- At 18 weeks, a detailed (fetal anomaly) scan can pick up a range of physical problems.
- Further testing may be necessary following this scan.
- Ultrasound does not harm the baby.

10½-13½ weeks
*FIRST TRIMESTER SCREENING USING NUCHAL TRANSLUCENCY ULTRASOUND WITH OR WITHOUT TESTING THE MOTHER'S BLOOD (*See Figure 3*)*
Screening test

Using ultrasound, the depth of fluid in the space at the back of the baby's neck is measured. This measurement is used to calculate the risk or chance of the baby having certain problems with chromosomes, such as Down syndrome.

- A special blood test measuring two proteins in the mother's blood taken at the same time as the ultrasound can give a more accurate risk result.
- Not all babies with a problem will get an increased risk result.
- This test does not give a definite result, only a risk of certain problems. Further testing may be necessary to clarify an increased risk result.
- These tests do not harm the baby

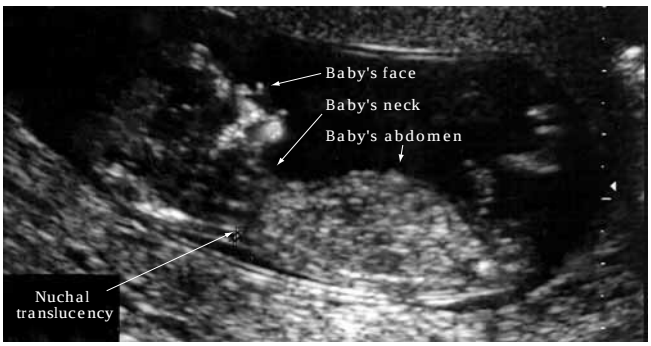

Figure 3: Nuchal Translucency

11-12 weeks
CHORIONIC VILLUS SAMPLING (CVS)
*(*See Figures 4 and 5*)*
Diagnostic test

During a CVS cells from the developing placenta are taken and tested for certain conditions such as Down syndrome.

- Chromosomes or DNA from the placenta is examined in the laboratory. Test results take about 1 week.
- There is a small risk (less than 1%) of miscarriage due to this test (in addition to the background risk of miscarriage in the first 3 months of pregnancy).

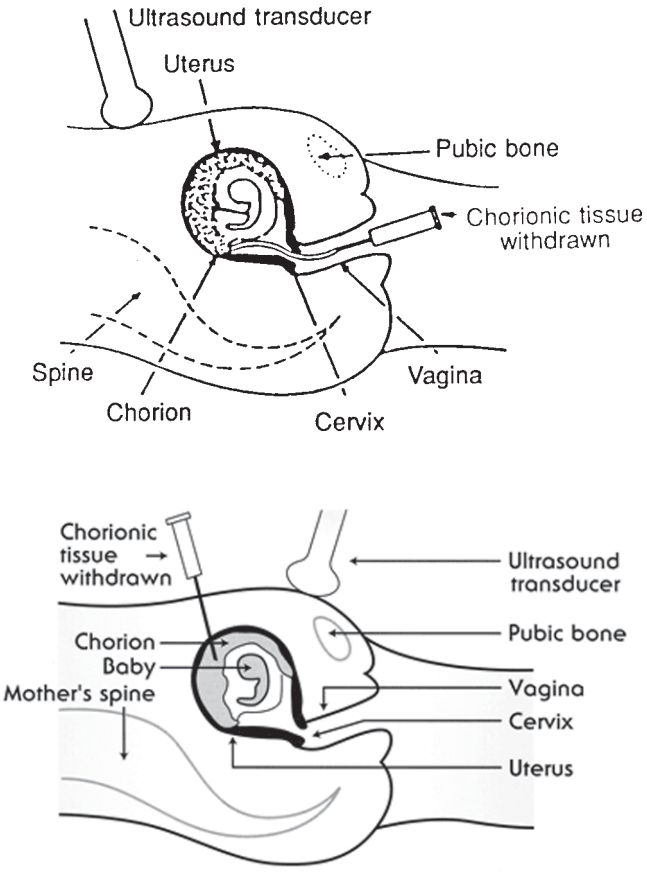

Figure 5. Abdominal CVS

15-17 weeks
MATERNAL SERUM TESTING (testing the mother's blood)
Screening test

Maternal serum testing looks at the mother's blood which does not give a definite answer but a risk that the baby has certain problems such as Down syndrome.

- It can also pick up most babies who have a neural tube defect such as spina bifida (see Genetics Fact Sheet 52), if used together with a detailed ultrasound scan.
- Not all babies with a problem will be detected.
- This test does not harm the baby.

15-19 weeks
*AMNIOCENTESIS (*See Figure 6*)*
Diagnostic test

Using a fine needle, a small amount of amniotic fluid is taken from around the baby and sent to the laboratory for analysis. Test results take between 1 and 2 weeks.

- The baby's cells are removed from the fluid and the chromosomes or DNA examined.
- The test will give a definite answer for problems such as Down syndrome.
- There is a slight risk of miscarriage (less than 1%) due to this test.

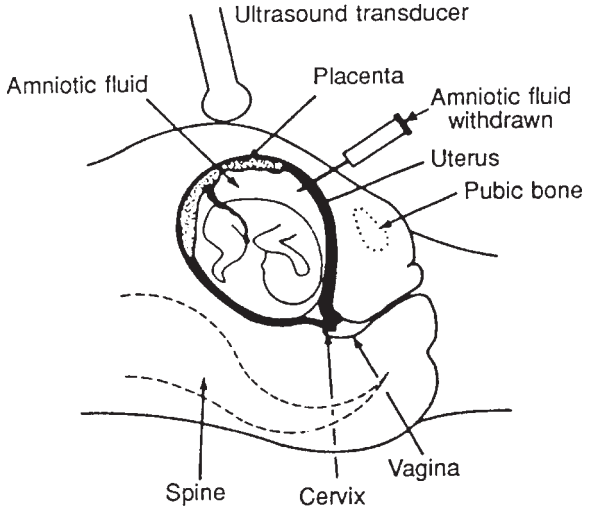

Figure 6: Amniocentesis

WHAT HAPPENS IF THE SCREENING TEST INDICATES THE BABY IS AT INCREASED RISK OF A CERTAIN PROBLEM?

An increased risk result does not mean that the baby definitely has a problem. Most babies who have an increased risk result will be normal.

It is important to discuss the result with a physician or genetic counsellor.

Further diagnostic testing may be indicated following an increased risk result on a screening test, in order to get a definite answer. This could be a CVS test, an amniocentesis test or a detailed ultrasound, depending on the length of the pregnancy.

Before deciding on any further testing, it is essential that the implications of the test are understood. All the options that may need to be considered following the diagnostic test result are best discussed prior to testing.

WHAT IF THE DIAGNOSTIC TEST SHOWS THAT THE BABY HAS A PROBLEM?

- It is important to obtain as much information as possible.
- Consider all available options.
- Support and counselling can assist in deciding what the most appropriate option will be.
- If the choice is to continue the pregnancy, support and information is important before, during and after the birth of the baby (see Genetics Fact Sheet 5: *Genetic Counselling*).
- If the choice is not to continue with the pregnancy, all aspects of the termination should be discussed, including the support available before and after the hospital stay.

THE IMPORTANCE OF COUNSELLING

Counselling provides an opportunity to discuss the advantages and disadvantages of having a screening or diagnostic test and any further testing that may be offered. It is recommended that these issues are discussed with a doctor or genetic counsellor prior to having any testing in pregnancy.

If a screening test result indicates that the baby has an increased chance of having a problem with his or her chromosomes, counselling is essential. This is an opportunity to discuss the advantages and disadvantages of further testing which could diagnose a chromosomal problem. Importantly, in most cases where an increased risk result following one of these screening tests has been found, the babies will not have a problem with their chromosomes at all.

Information in this Fact Sheet is sourced from:

Prenatal Diagnosis: Special tests for your baby during pregnancy (The Centre for Genetics Education)

Other Genetic Fact Sheets referred to in this Fact Sheet: 1, 2, 5, 12, 13, 16 and 52.