Fact sheet **26**

DIAGNOSTIC TESTS DURING PREGNANCY

This fact sheet talks about the types of tests available during pregnancy to diagnose certain genetic health and developmental problems in the baby.

IN SUMMARY

- Diagnostic tests in pregnancy look 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 genetic condition.
- A diagnostic test does not check for every possible condition which can cause differences in physical development or learning ability
- Prenatal diagnostic tests include:
  - Ultrasound
  - Chorionic villus sampling (CVS)
  - Amniocentesis (Amnio)

Pregnancy is divided into three stages (trimesters). The first trimester refers to the first three months of pregnancy, the second trimester spans the 4th to 6th months and the third trimester covers the final three months of pregnancy. Prenatal tests are done in either the first or second trimester of pregnancy.

PRENATAL DIAGNOSTIC TESTS FOR YOUR BABY IN PREGNANCY

There are a number of different tests available to assess the health and development of your baby before birth. On the following page is a **summary of the different diagnostic tests**, when they can be done, how reliable they are and also what information they provide.

It is important that you get as much information as possible before any testing is done. Talking about prenatal testing options should be offered to you and your partner in a safe and understanding way. Seeking support at this time may help you with 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 in the usual way or could develop a condition, having genetic counselling will give you the chance to talk about:

- 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 about disability
- Your thoughts and feelings towards termination of pregnancy (stopping/ending the pregnancy).

TYPES OF DIAGNOSTIC TESTS

First trimester ultrasound:

- Is done between 8-12 weeks of pregnancy
- Will confirm how many babies there are and how many weeks the pregnancy has progressed
- Is an important step if you are considering having testing later in your pregnancy

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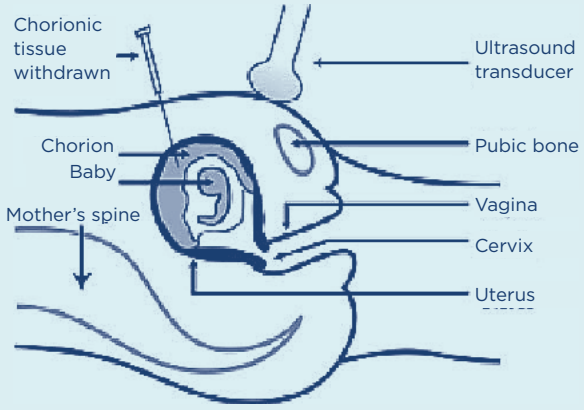
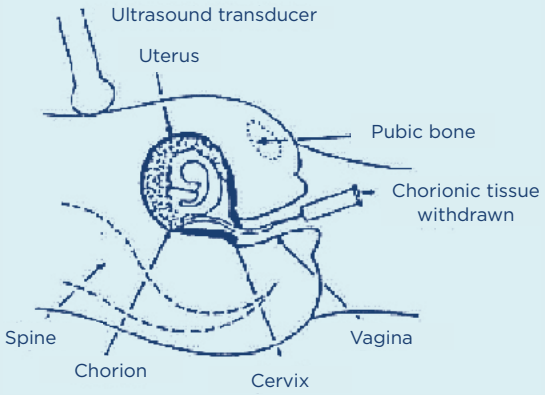
Figure 26.1:
Chorionic villus sampling in which about 10-20mg of chorionic villi are removed through the vagina and cervix usually done between 11-13 weeks of pregnancy. The procedure may be performed either trans-abdominally or trans-vaginally. Source: Vogel, F & Motulsky, A.G. (1986). Human Genetics 2nd ed. Springer-Verlag, Berlin

a) Trans-abdominal CVS



b) Trans-vaginal CVS



- May identify major problems with the growth of your baby
- Does not pose any health risk to you or your baby.

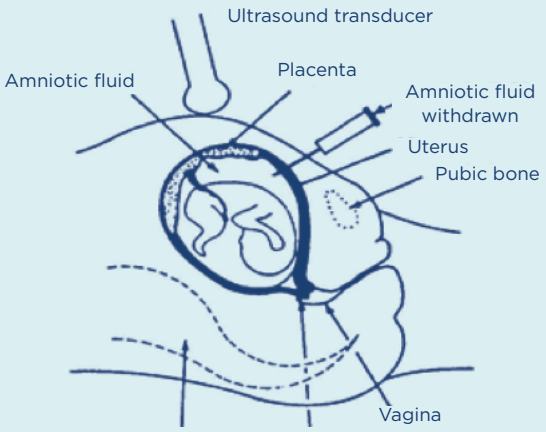
Chorionic villus sampling (CVS) (*Figure 26.1*):

- Is done between 11-13 weeks of pregnancy
- This test gives an accurate result for the genetic condition being tested for
- A sample of the chorion (placenta) is collected either using a needle through the abdomen or a narrow tube through the vagina and tested to determine if the baby has certain genetic conditions
- Depending on the type of test, results are usually available within two to three weeks
- Less than 1% of women (1 in 100) will have a miscarriage as a result of having a CVS.

Amniocentesis (*Figure 26.2*):

- Is done between 15-19 weeks of pregnancy
- This test gives an accurate result for the genetic condition being tested for
- A sample of the amniotic fluid surrounding the baby is collected and tested to determine if the baby has certain genetic conditions
- Depending on the type of test, results are usually available within two to three weeks
- Less than 1% of women (1 in 100) will have a miscarriage as a result of having an amniocentesis.

Figure 26.2:
Amniocentesis in which amniotic fluid (containing cells from the baby) is usually sampled between 15-19 weeks of pregnancy. Source: Vogel, F. & Motulsky, A.G. (1986). Human Genetics 2nd ed. Springer-Verlag, Berlin



Second trimester ultrasound:

- Is done between 18-20 weeks of pregnancy
- Is a safe way of checking your baby's growth and development
- In a small number of cases may identify that your baby has a genetic condition or some other health or developmental problem. Other follow-up tests may be offered
- Does not pose any health risk to you or your baby.

Other examples of pregnancy imaging if suggested by a doctor:

- MRI stands for Magnetic Resonance Imaging. This may be done in the second trimester of a pregnancy if a more detailed look at how the baby is developing may be needed. For example, images of the developing brain might show us more on MRI compared to an ultrasound at the same stage of pregnancy
- A heart scan for the pregnancy (also called a fetal echocardiogram or 'echo') is sometimes indicated. This may be done in the second trimester to safely take a good look at the different parts of the baby's heart to check for differences in how it formed and how it is working.

What might happen if a pregnancy sample is taken?

Sometimes a combination of diagnostic tests may be useful in your pregnancy. For example if an ultrasound shows a difference in the growth and/or development in the baby, this may be followed up with a detailed genetic test on a sample taken from the pregnancy (via CVS or amniocentesis). Further testing such as a chromosome microarray (looking for extra or missing sections of DNA) may indicate a genetic cause or a diagnosis of a health condition in the pregnancy.

In some situations your doctor might suggest a genomic testing of your pregnancy if for example differences are found on ultrasound. Genomic testing is where many genes can be tested for at the same time in one test.

In some instances this can increase the chance of finding a genetic reason for ultrasound differences in the pregnancy. A genetic diagnosis may also help us to better predict the likely nature of health or developmental problems for the baby beyond what can be seen on ultrasound.

The value of genomic testing may depend on the type of differences found on ultrasound and the likelihood that testing will provide a clear answer. Sometimes genomic testing may be less helpful. Your health care professional can further explain any limitations, benefits and disadvantages of tests available to you.

If a diagnostic test result shows that your baby has a health or developmental condition, you and your partner will be given as much support and information as possible about the condition and what this might mean for your pregnancy. You can also talk about whether any extra testing may give you a better understanding of what the results may mean for your pregnancy.

Your doctor, midwife or genetic counsellor can give you the opportunity to make an informed choice about whether or not you wish to have further testing and/or continue your pregnancy or not.

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