

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

In summary

- **Diagnostic tests 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 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).**

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.

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. Below is a summary of the different screening 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. 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 people with disabilities
- Your thoughts and feelings towards termination of 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
- *May* identify major problems with the growth of your baby
- Does not pose any health risk to you or your baby.

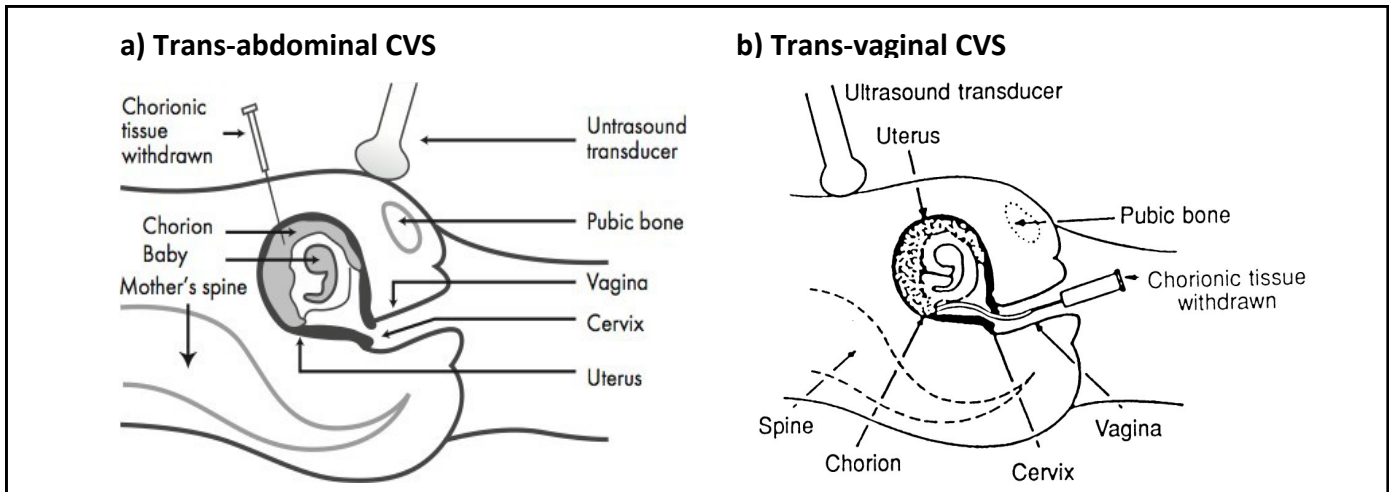


Figure 26.1: Chorionic villus sampling in which about 10-20mg of chorionic villi are removed through the vagina and cervix usually during the 11th to 12th week of pregnancy. The procedure may be performed either trans-abdominally or trans-vaginally. Source: Vogel, F & Motulsky, A.G. (1986). *Human Genetics 2nd ed.* Springer-Verlag, Berlin

Chorionic villus sampling (CVS) (Figures 26.1):

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

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
- Does not pose any health risk to you or your baby.

If a diagnostic test result shows that your baby has a problem, you and your partner will be given as much information as possible about the condition including the implications it might have for your baby's future health and development. You can also discuss whether any further testing may help you understand the situation.

You will be given time to make an informed choice about whether or not you wish have further testing and/or continue your pregnancy or not.

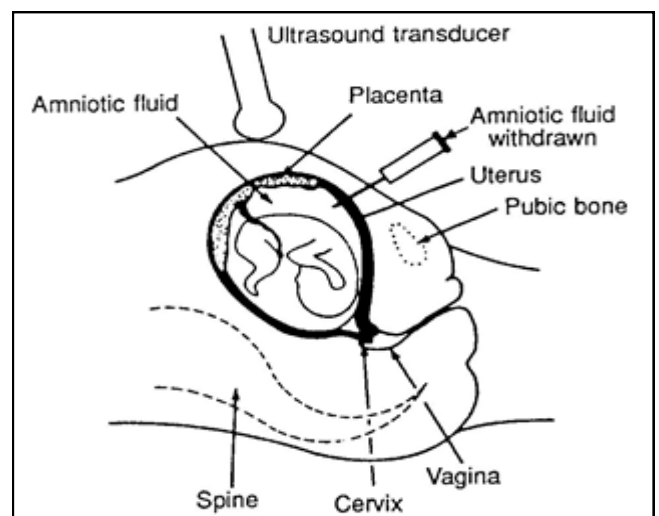


Figure 26.2: Amniocentesis in which amniotic fluid (containing cells from the baby) is sampled during the 15th-19th week of pregnancy. Source: Vogel, F. & Motulsky, A.G. (1986). *Human Genetics 2nd ed.* Springer-Verlag, Berlin.