

**TESTING YOUR  
BABY  
FOR GENETIC  
PROBLEMS  
DURING  
PREGNANCY**

**INFORMATION FOR PARENTS**

There are two main ways of checking your baby for genetic problems while it is in the womb. These techniques are called **amniocentesis** and **chronic villus sampling (CVS)**.

This leaflet explains how the tests are carried out, what sort of abnormalities can be picked up and what risks are involved.

---

The leaflet gives general information only. If you want more detailed information about these tests or your personal circumstances do discuss the matter with your GP or hospital specialist

---

### **What is amniocentesis?**

The amniocentesis test is usually carried out between 16 and 18 weeks of the pregnancy and can be carried out in an outpatients clinic without the need for an overnight hospital stay.

The test involves placing a needle in to the womb through the woman's abdomen (tummy) and obtaining a small sample of fluid in which the baby floats (the amniotic fluid). Most women say that this test is not painful.

As part of the test an ultrasound scan is carried out to check whether twins are present and to see where the placenta and the baby are.

The fluid contains cells, which have come from the baby, and these cells are then grown in the laboratory so that the genetic tests can be carried out on them.

### **What type of abnormality can be detected?**

Abnormalities caused by extra or missing chromosome material can be detected, the most common of which is down syndrome (mongolism), Where babies have an extra chromosome 21 in every cell of their body.

Other chromosomal abnormalities can also be detected. The effects of all these conditions will be discussed in detail with the parents. Some conditions are at least as serious as Down syndrome and a termination of pregnancy may be one option for the parents to consider.

In some cases, other disorders caused by abnormal genes can be found. At present tests for these are usually carried out when there is someone with the condition in the family of the couple is known to be at risk for a baby with particular genetic diseases, such as cystic fibrosis, sickle cell anaemia or Tay Sachs Disease.

The amniotic fluid itself is also tested to measure the level of a protein called alpha-fetoprotein (AFP). The level is high if a baby has spina bifida, and this test picks up 95% of babies with this disorder. A detailed ultrasound scan is then carried out to confirm the presence of spina bifida.

### **What are the risks of the test, and how reliable is it?**

The amniocentesis test carries a small risk causing a miscarriage than chorionic villus sampling but it has the advantage of being able to be done earlier in the pregnancy.

The amniocentesis test is a very reliable way of examining the baby's chromosomes, but in order to obtain results the cells must begin to grow in the laboratory. Occasionally this does not happen and a further sample may be needed.

### **When is the result available?**

Within three or four weeks if an abnormal result is found and the couple decides to have the pregnancy terminated, this could be done at around 19-20 weeks of the pregnancy.

### **What is chorionic villus sampling (cvs)?**

The CVS test is carried out earlier than amniocentesis – at around the 9<sup>th</sup> or 10<sup>th</sup> week of the pregnancy and again it is an outpatient procedure. Rather than testing and sampling of the fluid of the mother's womb, this test takes a very small amount of material from the developing placenta.

One way of doing the test is to place a thin tube through the cervix (neck of the womb) to obtain the sample. Most women say that this is only slightly uncomfortable.

Another method is similar to the amniocentesis test, and involves placing a needle in the womb and using an ultrasound scanner to guide it.

### **What type of abnormality can be detected?**

The sample contains cells that are of the same genetic type as the baby and so can reveal the same sort of disorders that amniocentesis discovers. Similarly CVS can detect rare diseases caused by abnormal genes, but these will only normally be done if there is a family link with a particular disorder. Because no amniotic fluid is obtained the test cannot be used to look for babies with spina bifida.

### **What are the risks of the test, and how reliable is it?**

The CVS test has a slightly higher chance of a miss carriage than amniocentesis but it has the advantage of being able to be done earlier in pregnancy.

It is relatively new test and there is not so much information on reliabilities with amniocentesis. There is a suggestion that the CVS test may give a confusing result in proportion of cases, maybe one in 100.

In such case it may indicate an abnormality where none exists. Although there is usually additional information, which clearly demonstrates that the baby will be abnormal, where doubt still exists it, may be necessary for an amniocentesis test also to be carried out later in the pregnancy

There is also a remote possibility – around 1 in 1000 cases- that chromosome normality may not be detected.

### **When is the test available?**

Normally just one to two weeks after the test.

### **What about future pregnancies?**

If a couple decide to terminate their pregnancy after either an amniocentesis or a CVS test , this should not affect their fertility future pregnancies. The couple may well wish to be referred to their genetic clinic to discuss the risk of an affected baby next time.

### **Rhesus negative mothers**

If a mother's blood group is rhesus negative, she should receive an injection after an amniocentesis or CVS to protect the present and future pregnancies

Published by the Specialist Group In Clinical Genetic  
Northwest Thames Regional Health Authority  
Reproduced by the Sickle Cell Society – April 98

