



**SICKLE CELL  
FOUNDATION NIGERIA**  
Bringing Hope to the African Child

## **PRENATAL DIAGNOSIS (WHAT WHERE AND WHEN)**

This is a test that can be done on the baby before it is born to see whether it is affected by a particular disease or not. If it is affected, the couple may decide to terminate the pregnancy and try again for an unaffected child or may choose to continue with the pregnancy and prepare their minds for its birth. If the baby is not affected, then the pregnancy can continue normally.

Remember that, for most couples at risk for sickle cell anaemia (e.g. AS x AS) there is a 3 out of 4 chance of a healthy child in each pregnancy continue normally after the test.

### **The Different Tests Available**

Material from the baby can be obtained for testing in three different ways, called

- (i) **Chorionic Villus Sampling,**
- (ii) **Fetal Blood Sampling**
- (iii) **Amniocentesis.**

The type of test done depends on the stage of pregnancy and the position of the placenta. The test can be done between 10 and 22 weeks of pregnancy, but obviously the earlier, the better.

An early test is less upsetting than a later test, because if they wish to termination is easier and more acceptable than a later one.

Nowadays the best way to examine the material from the baby is by analyzing the haemoglobin genes. This is called "DNA analysis". Using this method it is possible to detect the genes for haemoglobin A and haemoglobin S in the material from the baby's placenta.

## **Chorionic Villus Sampling (CVS)**

This test can be done early in pregnancy from about 8 weeks after the last timing is between 10 and 14 weeks.

CVS is performed by a trained obstetrician using 2 possible routes:- transcervical or transabdominal, both under continuous ultrasound guidance. The route chosen depends on the site of the placenta and the preference of the sampler.

No anaesthetic is required except a local one for the transabdominal approach. Clients can get up and leave a few minutes after the sample is obtained. They should take things easy for a few days afterwards to minimize risk of miscarriage.

### **HOW THE CVS IS TESTED.**

DNA from the chorionic villi is studied to see if the baby's genes for haemoglobin are normal, or if an alteration has been passed on from the parents. It takes from 3 to 10 days to analyze DNA, so we usually have the result in about one week.

### **Is The DNA Test Accurate?**

DNA analysis is the best method for diagnosing inherited disorders. However in every medical test there is a small possibility of every medical test there is a small possibility of an error. The chance of a mistake is very small; less than 1 in 200 (0.5%).

### **Is CVS Safe?**

There is practically no risk to the mother. The main risk is that the test could cause a miscarriage. We do the test as gently and expertly as possible, but anything that interferes with a pregnancy can cause a miscarriage. At the moment, the risk of miscarriage after CVS during the first 12 weeks seems to be around 1 in 50 (2%).

However it is difficult to give an exact figure because other factors may influence the risk of miscarriage after this test.

For example, the older the mother the higher the risk of miscarriage. When a miscarriage does occur, it can be difficult to tell whether it was due to the test or not, because many miscarriages happen naturally at around 12 weeks of pregnancy without any interference.

### **Fetal Blood Sampling**

This can only be done from 18-22 weeks after the mother's last menstrual period. The needle for obtaining the blood is passed through the mother's abdomen to the baby's umbilical cord without touching the baby. As usual the procedure is guided all the way by an ultrasound scanner.

After the test the women can go home after resting in hospital for about 30 minutes. The results are very reliable and can be obtained by DNA analysis or by analyzing the baby's haemoglobin. The risks of this procedure are very small. None practically to the mother, while in about 1 in 100 cases a miscarriage might occur.

\*Fetal blood sampling is principally used either when the pregnancy is already far advanced or the placenta cannot be reached for CVS or when DNA analysis is not possible.

### **Amniocentesis**

Technically it is the easiest to perform of the 3 tests described here but because of the long process required in the laboratory before DNA analysis can be done, it is used only rarely when for some reason CVS or fetal blood sampling cannot be done.

It carries less than a 1 in 100 risk of miscarriage and virtually no risk at all to the mother. The result takes a minimum period of 3 weeks.

### **When Was Prenatal Diagnosis Introduced To Nigeria**

Prenatal diagnosis was introduced to Nigeria in November 1993 through an initiative of the Sickle Cell Club Lagos Nigeria and the collaboration of the British Council, the department of Obstetrics and Gynecology of University Teaching Hospital and the Nigerian Institute<sup>4</sup> of Medical Research.

## **WHAT THE CLIENT SHOULD EXPECT**

### **Reception and Counseling**

Our receptionist at the College of Medicine will make an appointment for the couple to give a blood sample and meet our trained Sickle Cell Counselor who will note the date of the last menstrual period the obstetric and medical history and ensure that they understand all the relevant information.

### **Preliminary Tests**

The blood of husband and wife will be drawn in the laboratory for some preliminary tests and the wife will be given a date for meeting the obstetrician who will perform an ultrasound scan to inspect the fetus. If the partner cannot come himself, the woman can collect a specimen tube to our laboratory.

**For further information, please contact;**

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