after CVS during the first 12 weeks in experienced hands is less than 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. One factor is the age of the mother. Older mothers have a higher risk of miscarriage. When a miscarriage occurs, it can be difficult to tell whether it is due to the test or not, because many miscarriages happen naturally at around 12 weeks of pregnancy without any interference.

Very rarely, in about 6 out of 100,000 cases, CVS can cause shortening of development of the limbs or fingers of the baby. However, this has only happened if the test was done around 8 weeks of pregnancy. There is no evidence that CVS lead to any such problems when it is done after 9 weeks of pregnancy.

AMNIOCENTESIS

This word means the withdrawal by syringe and needle of a sample of the fluid surrounding the fetus or unborn baby. 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 cannot be done. It carries less than a 1 in 100 risk in miscarriage and virtually no risk at all to the mother. The result takes a minimum period of 3 weeks.

FETAL BLOOD SAMPLING

In this test, a blood sample is taken from the fetus. 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 tummy 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 mother can go home after resting in the hospital for about 30minutes. It takes about a week to get the result.

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 with the collaboration of the British Council, the Departments of Obstetrics and Gynaecology of University College London and of the Lagos University Teaching Hospital and the Nigerian Institute of Medical Research.

WHERE IS PND AVAILABLE IN NIGERIA?

So far, only in Lagos until the necessary skills and facilities are extended to other towns. Those who would like to undergo the test should contact the National Sickle Cell Centre, Ishaga Road, Idi-Araba, opposite the Lagos University Teaching Hospital. Tel: (01) 725 3957-8; 0803 319 5177.

WHAT YOU SHOULD EXPECT

Reception and Counselling

Our receptionist will make an appointment for you to give a blood sample and meet our counsellor who will note the date of your last menstrual period (LMP), your medical history and ensure that you understand all the relevant information, including the prescribed fee.

The Fee for PND

Considering the organisational, administrative, operative and laboratory procedures and materials, and the time and skills of professionals involved at every stage, PND is not cheap. We need a subsidy to make it affordable to most Nigerians who want it and we are seeking such sponsorship.

Preliminary Tests

The blood of your partner and yourself will be drawn in the laboratory for some preliminary tests and you will be given a date for meeting the obstetrician who will perform an ultrasound scan to inspect the fetus. If your partner cannot come himself, you can collect a specimen tube and get him to find a doctor to collect 3-5ml of his blood into the tube and bring it along with you.

ULTRASOUND EXAMINATION AND CVS

The ultrasound scan does not involve any invasion of any part of your body and is completely painless and harmless. The scan will enable the obstetrician to correctly assess the size of your baby (your dates maybe wrong!), see whether you are carrying twins or more, and note the location of the placenta. After which the CVS may be taken straightaway or you are given the best date for doing so in the interest of the safety of the baby. You should return to see the counsellor for the result about 10 days after the test. Check the date before you go home. In the event that the result is genotype Hb SS, you should discuss the options with our counsellor including early preventive care of the baby after birth. We shall be pleased to keep in touch with you thereafter and provide the required support.

In the unlikely event that you lose the pregnancy or experience any bleeding, you should call to see our obstetricians or if more convenient your own doctor, but please keep us informed.

FINALLY

We wish you a safe delivery. You should bring the baby back to see us 6 months after delivery for examination and blood test to confirm the prenatal diagnosis result.

Issued by:

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

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SICKLE CELL PRENATAL DIAGNOSIS

What You Need to Know

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

WHAT IS PRENATAL DIAGNOSIS?

Prenatal diagnosis (PND) is a diagnosis made in an unborn child (foetus or fetus) still in the mother's womb. PND is usually requested by a couple who have a risk of bearing children with a particular disorder, usually an inherited one, such as sickle cell anaemia.

WHY CHOOSE PND OF SICKLE CELL DISORDER?

It is worth noting that most couples who are at risk of bearing children affected with sickle cell anaemia (Hb SS) are themselves healthy carriers of the sickle cell gene (i.e. both would have Hb AS) and thus have 3 out of 4 chances (a 75% chance) in each pregnancy, of bearing an unaffected child (i.e. with Hb AA or AS). Many such couples cherish the opportunity to know the child's diagnosis before birth, in order to relieve anxiety that the child is not affected or to prepare their minds for the birth, if affected.

Depending on circumstances, such as whether or not they already have affected children and their perception of the quality of care that would be available to an affected child, the couple may decide to continue with the pregnancy and prepare their minds for the birth of the baby or choose to terminate the pregnancy bearing an affected child and try again for an unaffected child. Now that better health care of affected children is available and is increasing their life expectancies and reducing their frequency of illness, more and more couples are choosing to continue with affected pregnancies.

THE DIFFERENT PND TESTS AVAILABLE

The 3 possible procedures are (i) **chorionic villus sampling**, (ii) **amniocentesis** and (iii) **fetal blood sampling**.

The recommended procedure for inherited haemoglobin disorders is chorionic villus sampling (CVS) as it can be carried out early in pregnancy and with a high degree of safety in experienced hands. Fetal blood sampling is only possible quite late in pregnancy and requires such highly specialised equipment and expertise that it is hardly used anywhere now for PND of sickle cell disorder. Amniocentesis is less difficult but cannot be done in the first three months of pregnancy or diagnosis obtained as rapidly as with CVS and so it is less favoured.

The modern method for diagnosing the sample from the baby is by DNA analysis of the haemoglobin genes.

CHOOSING PRENATAL DIAGNOSIS

Once you have become pregnant, we ask you to call our Centre as soon as possible for an appointment to see us. If possible, it is important for both partners to come for this first visit. The first visit is necessary for the following reasons.

1. To counsel you and fully discuss the advantages and risks of the procedure, answer your questions, and provide all the information you would require in making your decision.
2. To take a blood sample from you and your partner, if you decide to go ahead.
3. To prepare your medical records.

If you choose to have prenatal diagnosis, you make a separately appointed visit for the procedure. You come to the Centre alone or with your partner. We do an ultrasound scan to see exactly where the baby and the placenta are. During the procedure to obtain the sample for diagnosis, we use the ultrasound scan throughout so that we can see exactly what is happening. You are not put to sleep and you can go home later the same day.

CHORIONIC VILLUS SAMPLING

CVS can be safely done early in pregnancy from about 9½ weeks after your last menstrual period. The best timing is between 10 and 14 weeks.

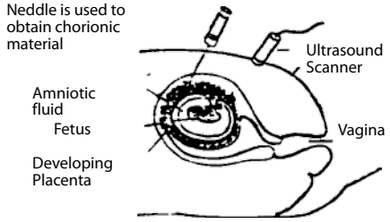
A small amount of material is taken from the developing placenta. The placenta is where the baby is attached to the mother. It develops from the tissues of the baby, not of the mother, so it has the same genes as the baby. It is made of up of chorionic villi. Villi is plural for villus.

Two routes may be taken to obtain a sample of chorionic villi; either through the abdomen (tummy) or through the cervix of the uterus via the vagina. The abdominal route is mostly taken except when the position of the placenta dictates otherwise. Most samples are therefore obtained through the abdomen and this route leads to a much lower chance of a miscarriage of the pregnancy than going through the vagina and cervix.

The procedure is carried out by a specially trained and experienced obstetrician. We use an ultrasound scan all the time so that we can see what we are doing.

We reach the placenta by putting a long needle through the abdomen and into the womb. We inject a local anaesthetic into the skin in order to prevent pain before we insert the needle. The local anaesthetic stings a little and there is a brief feeling of pressure when the needle is put in, but you should feel very little pain. When the ultrasound picture shows that the needle is in the right place, the obstetrician fixes a syringe to it and gently removes some chorionic villi.

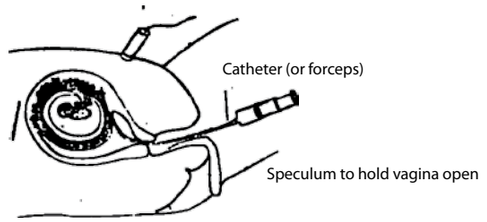
Alternately, the obstetrician puts a very thin forceps through the vagina and then into the womb. It is so thin that



most women hardly feel it. It does not touch the baby, or the little bag of water surrounding the baby. Then the forceps are opened and closed to take a very small sample of chorionic villi from the placenta. Alternatively, the obstetrician may pass a very thin tube by the same route to the placenta. Then a syringe is attached to the outer end of the tube and a very sample of the chorionic villi is sucked out. The picture shows the route.

Once the sample has been obtained, we immediately examine it under a microscope to check that it is the right sample from the placenta. If it is, we stop. If it is not the correct sample, we manipulate the tip of the tube or needle slightly to obtain the right sample. The test usually takes 10 t0 20 minutes.

After the test, we invite you to lie down for about 30 minutes before you go home. It is wise to take things easy for a



day or two. This means you can go about as usual, but should avoid long journeys for a few days and sexual intercourse for about 10 days after the test. If the test has been done through the vagina you may observe some blood spots for a few days. This is usually from the vagina and not the womb and is usually harmless. However, if there is a lot of blood or you have pain or fever, you should contact us immediately.

HOW THE CVS IS TESTED

The genes responsible for all the characteristics inherited from parents, including haemoglobin, are made of a material called DNA. All the tissues of our body contain our whole DNA pattern. In the fetus this includes the placenta. We study DNA from the chorionic villi to see if the baby's genes for haemoglobin are normal, or if an alteration has been passed on from the parents. It takes a few days to analyse DNA, so we usually have the result in about one week, or longer, when we have to send it abroad for analysis.

IS DNA TEST ACCURATE?

DNA analysis is the best method for diagnosing inherited disorders. However, in every medical test there is a small possibility of an error sometimes, arising from human error but sometimes due to nature 'playing a trick'. 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 lead to a miscarriage. We do the test as expertly as possible but anything that interferes with a pregnancy can cause a miscarriage. At the moment, the risk of miscarriage