



PRENATAL TESTING

ULTRASOUND

A variety of prenatal tests are available. While not all patients need or will have all the various tests, it is good to know that they are available. These tests have different purposes, but they all share a common goal: monitoring the health of both the fetus and the expectant mother. In addition to this material, you should also read about alpha-fetoprotein testing and other routine blood testing for pregnancy.

Ultrasound is used with other tests to determine the location of the fetus, its gestational age, and to guide the medical instruments used in other prenatal tests. This test can, however, be used by itself to detect problems with the fetus and placenta, as well as the number of fetuses in the womb. At 18 to 20 weeks into the pregnancy, facial features, organs, and limbs can be seen by a skilled technician. For example, certain facial features can indicate that the baby might be born with Down's syndrome. In many cases, though, the results of an ultrasound are not absolute; women must undergo more definitive tests.

Although ultrasound can aid in the detection of many birth defects, many doctors dispute the screening of all babies. It has not been proven that ultrasound produces healthier babies, but no one can deny its usefulness when performed.

The results of an ultrasound can be seen immediately; this creates both advantages as well as disadvantages. Since an actual picture is produced, allowing the mother to see her fetus, bonding between mother and "child" can begin early on in pregnancy. Bonding increases dramatically further along in the pregnancy, especially when the development of genitals and other "person-like" features can be seen on the ultrasound picture (fingers, a nose, feet, etc.).

Ultrasound can often make a pregnancy fun by giving the parents a sneak preview of what is to come, but the joy can end quite abruptly if a problem is found. Because results are obtained immediately, and abnormalities seen instantly, the parents may be unprepared to handle disappointing news especially when they are not expecting it.

AMNIOCENTESIS

One of the more common tests in high-risk pregnancies is amniocentesis. During amniocentesis, a needle is inserted through the abdomen into the amniotic sac where the embryo grows into a fetus, and several teaspoons of fluid, called amniotic fluid, is withdrawn into a syringe at the end of the needle. The fluid is analyzed in a laboratory and results are ready in 2 to 3 weeks.

This test, however, is not suitable for all pregnant women. Because of the nature of the procedure, there is the small possibility of a miscarriage. This risk is the reason that amniocentesis is usually offered only to those women with a high risk of fetal abnormality due to age or family history.

Amniocentesis can detect certain metabolic disorders and chromosomal abnormalities such as Down's syndrome, Tay-Sachs disease, and neural tube disorders such as an opening in the spine. Results of amniocentesis are much more accurate than ultrasound, and therefore should be regarded more highly.

CHORIONIC VILLUS SAMPLING (CVS)

Chorionic villus sampling, or CVS, is a test that is performed between the 12th and 14th week of pregnancy. This procedure involves obtaining cells from tiny hair-like projections, called villi, from the chorion surrounding the embryo. CVS is accomplished with the use of ultrasound equipment and a catheter that is inserted into the vagina, through the cervix, enabling the cells to be taken.

Another method, called transabdominal CVS, has the catheter passed through the abdomen into the uterus instead of the vagina and cervix. There is not, however, a predominant method; there is no research that shows a better method between the two.

The positive aspects of CVS lie in the ease with which it is administered. Cells taken from the villi can be analyzed directly, without preparing a culture; this enables doctors to examine fresh cells, without wasting time. Early results from CVS can be obtained within 48 hours, and final results may be obtained about 10 days later. Early results mean early diagnosis for genetic defects such as Down's syndrome. They also make the waiting period bearable for expectant mothers.

The negative side of chorionic villus sampling lies in its accuracy and risks. The main reason that the cells that are examined are taken from the chorion is that the tissue of the chorion is very similar to that of the fetus. The error in CVS stems from the fact that some abnormalities in the chorionic cells--those that are examined--do not occur in the fetus. Because of this, obtaining false positive results has led many women to abort a normal baby. This especially occurs in the diagnosis of mosaicisms, or abnormalities that occur in some cells but not all of them. In the case of mosaicism, doctors order an amniocentesis to confirm results of CVS. Since the test involves penetration of the vagina and cervix, mothers run the risk of contracting infections through the cervix.

FETOSCOPY

Fetoscopy, developed in the 1950s, was used to visualize the fetus and detect certain hereditary diseases that couldn't be seen in the analysis of amniotic fluid. This procedure was also used for fetal blood sampling, in which case the instruments would be used to visualize fetal blood vessels.

In fetoscopy, doctors would use ultrasound equipment and other instruments in order to take tissue samples from the skin, liver, or muscles of the fetus. These

tissues would be analyzed, and the doctors would check for certain diseases such as hemophilia--hereditary blood disorders that could only be seen in the actual DNA.

FETAL BLOOD SAMPLING AND PERCUTANEOUS UMBILICAL BLOOD SAMPLING (PUBS)

Fetal blood sampling, however, is a procedure that is becoming obsolete. New technology has made possible an improved screening technique called percutaneous umbilical blood sampling (PUBS). These procedures take less time than fetoscopy, requires less physical strain on the mother, and the results are available much faster.

PUBS is performed in basically the same way as fetal blood sampling, but it is much more convenient for the mother. PUBS is performed at the same time as amniocentesis. A needle is passed into the large vein in the umbilical cord, and blood is drawn and studied. Results are available within 3 days, at which time the mother will find out if her baby has AIDS, sickle-cell anemia, or any other blood disease.

PUBS is useful not only for diagnosis, but also for some forms of treatment. This form of blood testing is done in a blood vessel that leads directly to the uterus. What this means is that if doctors can insert a needle to take blood out, they must also be able to inject certain liquid solutions at the same point. Medication can be given and, in some cases, blood transfusions can be performed--these are examples of treatment begun in utero. This makes it easier for doctors to begin treatment on the baby once it is delivered, because if physicians did not have this "head start," chances of survival would be much less.

While this form of blood sampling has many positives, there are many negative aspects as well. PUBS is difficult to perform, due to the fact that the umbilical cord moves around a lot. Second, the blood is very often contaminated with amniotic fluid, making it difficult to obtain definite results. There is a great risk of infection because of where the needle is being put in the mother's body, and because of accidents resulting in the penetration of the uterus, which can lead to a miscarriage. Statistics tell us that the rate of miscarriage for those mothers who have undergone PUBS is about 2% higher than normal.

FETAL BIOPSY

There is one other test that requires the use of fetoscopy: fetal biopsy. In this procedure, the instruments used in PUBS are used to penetrate the mother's abdomen into her uterus, taking samples of fetal skin, liver, and muscle tissues. With these samples, doctors can diagnose certain disorders not even obtained by DNA analysis, such as albinism from the skin sample. Like PUBS, the test results are available relatively quickly. Unfortunately, the rate of fetal loss as a result of fetal biopsy is rather high.

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