Prenatal Testing Options

During your pregnancy you may be given the option to have any one of several prenatal tests. Whether or not to have prenatal testing during a pregnancy is a very personal decision. It may be right for some people for certain reasons and not right for others. By knowing what is available, you can make a decision about testing that is right for you and your family. It is important to remember that prenatal testing is optional, and that you may elect to have no prenatal testing at all. If you choose to have prenatal testing, you may choose any combination of tests based on what is right for you individually. Prenatal testing can be divided into two categories, screening tests and diagnostic tests.

Screening tests

Screening tests do just that; they screen a pregnancy to determine whether the fetus is at risk for an abnormality. Screening tests were originally developed for women who were at a "low risk" for having a baby with a chromosome problem and would not otherwise be offered more invasive testing. Now even women in "high risk" groups are using screening tests to try to avoid more invasive testing. However screening tests may not be able to provide "high risk" women with as much reassurance. A "high risk" test result does not mean the baby is affected. In fact, most women with "high risk" results have normal follow-up tests and healthy babies. If a pregnancy is considered "high risk" you may choose to have a diagnostic test such as amniocentesis or cvs for further evaluation. If a pregnancy is considered at "low risk" based on the results of a screening test, it means that the risk is low enough that diagnostic testing is not indicated. The available screening tests include:

1st trimester Down syndrome screening (aka BUN screening): First trimester screening is designed for women interested in determining whether their baby is at increased risk for Down syndrome and Trisomy 18 (a more rare yet more severe chromosome abnormality). 1st trimester screening is done between 11 and 14 weeks gestation and can only be done on a singleton pregnancy. The screening is able to determine the risk of Down syndrome and Trisomy 18 with a 90% detection rate. The screening involves a blood test to measure blood protein levels and an ultrasound to measure nuchal translucency or the thickness of the skin on the back of the fetal neck.

Maternal Serum Alpha fetoprotein Plus screening: A maternal blood test done between 15 to 19 weeks of pregnancy to measure the amount of alpha fetoprotein and other chemicals which can determine the risk for Down syndrome and Trisomy 18 as well as open spinal column defects. This screening has a 60% detection rate for Down syndrome and Trisomy 18. The detection rate for open spinal column defects is up to 90%.

Level II ultrasound: A comprehensive or level II ultrasound is a thorough evaluation of the fetal anatomy targeted to identify anatomic or structural birth defects. Anatomic birth defects are present in 2-3% of liveborn infants. Fetal growth, amniotic fluid volume and placental anatomy are also evaluated during a comprehensive ultrasound examination. Chromosomal abnormalities cannot be detected or ruled out by ultrasound. However, fetal anatomic abnormalities are observed in 50-70% of fetuses with Down syndrome and in over 90% of fetuses with trisomy 13 or 18 (the next most common chromosomal abnormalities). Therefore, a normal comprehensive ultrasound may be associated with a reduced risk for chromosomal abnormalities. In contrast,

identification of fetal anatomic abnormalities on ultrasound may be associated with a higher risk of fetal chromosomal abnormalities.

With any of these screening tests, if an increased risk of abnormality in the fetus is determined, genetic testing, such as amniocentesis or chorionic villus sampling, may be offered for definitive diagnosis of fetal chromosomes.

Diagnostic tests

Developed for pregnancies at "high risk" for a chromosome abnormality, diagnostic testing can actually determine whether the fetus is affected or not. Women considered at "high risk" to have a baby with a chromosome problem include women who will be 35 years old or older at the time of delivery or women who have had an abnormal screening test. Diagnostic tests include:

Amniocentesis: A technique that is offered for prenatal diagnosis of chromosomal abnormalities. It is usually performed in the second trimester of pregnancy. During the procedure, a small sample of amniotic fluid is removed under ultrasound guidance. The fetal cells floating in the amniotic fluid can be used to do chromosomal studies. Results are available approximately 2-3 weeks after the amniocentesis is performed. The increase in the miscarriage rate following this procedure is thought to be 1 in 200 procedures or approximately 0.5% above the background risk for miscarriage during the 2nd trimester. Abnormalities in chromosomal number (more or less than 46) or in structure (an extra or missing piece) can be determined by amniocentesis analysis. Fetal sex can also be determined. Additionally, the amniotic fluid can be tested to measure the amount of alpha feto-protein which is a diagnostic test for detecting a birth defect called spina bifida (open defect of the spine).

Chorionic villus sampling (CVS): A technique that is offered for prenatal diagnosis of chromosomal abnormalities as an alternative to amniocentesis. CVS is performed between 10-13 weeks of pregnancy. During the procedure a small sample of the placenta, called the chorion, is removed under ultrasound guidance. The cells from the chorion can be used to do chromosomal studies. Results are available approximately 1-2 weeks after the CVS is performed. The main advantage of CVS over amniocentesis is that prenatal diagnosis is achieved during the first trimester which allows a couple the opportunity to consider their available options earlier in the pregnancy in the event of an abnormal result. The increase in the miscarriage rate following this procedure is thought to be 1 in 200 procedures or approximately 0.5% above the background risk for miscarriage, which is 2-3% at 10-12 weeks of pregnancy. Abnormalities in chromosomal number (more or less than 46) or in structure (an extra or missing piece) can be determined by CVS. Screening for neural tube defects, such as spina bifida, cannot be achieved through CVS. Patients who opt for CVS should be offered the blood test maternal serum alpha-fetoprotein, performed at approximately 16 weeks gestation to screen for neural tube defects.

Although amniocentesis and chorionic villus sampling test for chromosome abnormalities, a normal test does not guarantee that the baby will be free of all birth defects. Only 10 to 15 percent of birth defects involve abnormal chromosomes. For example, anatomic or structural abnormalities, such as heart defects, often occur in the absence of chromosomal abnormalities. Therefore, a normal test does not guarantee a healthy baby; it just means the baby will be free of the specific abnormality evaluated (chromosomal abnormalities).

For more information about prenatal testing please feel free to consult your healthcare provider or call the genetic counseling office at (847) 570-2864.