PRENATAL DIAGNOSTIC TESTING

This handout is designed to help you understand the available options and timing of prenatal testing for you and your baby. It is important to note that this testing is not required and will only be done when you and your provider decide together whether it is appropriate. You should only have these tests done if you want to know before birth if your child is affected by these diseases, or if that information will help you with your decision-making in this pregnancy. More information on each of these diseases is available in your *Planning Your Pregnancy and Birth* book and in disease-specific handouts available upon request. It is important that you know we do not have tests for all conditions that can affect babies, and that no test guarantees a perfect outcome in pregnancy.

CYSTIC FIBROSIS TESTING:

This is a blood test offered to all mothers to see if they carry the gene for Cystic Fibrosis (CF) and it can be done at anytime before or during pregnancy. If the mother is a gene carrier, it is then necessary to test the father's blood. If both parents carry the gene for CF, the fetus is at risk for the disease. CF causes the body to make large amounts of thick mucus in the lungs and intestines. Today, there is no cure for CF and the illness can lead to frequent hospitalizations. People with CF usually live between 30 and 50 years. The incidence of CF by ethnic groups is the following: Caucasians 1 in 3,300, Hispanic 1 in 8,000-9,000, African American 1 in 15,300, and Asian Americans 1 in 32,100. Some insurance companies do not cover the cost of this test, which is approximately *$265*. ?

SPINAL MUSCULAR ATROPHY TESTING:

This is a blood test to see if you are a carrier of the Spinal Muscular Atrophy (SMA) gene, which can be done at anytime before or during pregnancy. If the mother is a gene carrier, it is necessary to test the father's blood. If both parents carry the gene for SMA, there is a 1-in-4 chance with each pregnancy that the child will be affected. SMA is a hereditary disease that destroys the nerves responsible for controlling voluntary muscle movement, but does not affect intelligence. Muscles that control breathing, swallowing, head and neck control, walking and crawling are the most severely affected. Most symptoms show up before a baby is two years old. There is currently no cure or treatment for SMA. Insurance usually covers this test if you are currently pregnant. The cost is $472.

FIRST TRIMESTER BIRTH DEFECT SCREENING:

This test is available to all women. Insurance usually covers this test. First trimester screening is based on measurement of two maternal serum proteins (PAPP-A and beta hCG), nuchal translucency, and maternal age to calculate the risk for Down Syndrome and Trisomy 18. This test is non-invasive and requires maternal blood work and an ultrasound of the fetus. First trimester screening detects 80-85% of Down Syndrome and 90% of Trisomy 18 babies. Because this test is done between weeks 11-14, it allows time for early diagnosis of these birth defects. This test is slightly more accurate than the traditional Quad Screen offered to women at 16-18 weeks. We do not offer this test in the office but can refer you to a nearby testing site.

CVS OR CHORIONIC VILLUS SAMPLING:

This testing is offered only to women over age 35 and women with specific high risk factors. Insurance usually covers this test. This procedure involves taking a sample from part of the developing placenta, the chorionic villi. These chorionic villi cells have the same genetic make up as the fetus and can detect Down Syndrome, and Trisomy 13 and 18. The benefit of this test is that it is done at an earlier gestation (12 weeks) than the amniocentesis but the risk of the

procedure is more significant. Your provider will help you decide whether this procedure is appropriate for you.

QUAD SCREEN:

This test is offered to all women and is usually covered by insurance. The Quad Screen is a blood test done on the mother at 16-18 weeks of pregnancy. Testing tells us whether your pregnancy is at higher risk for Down Syndrome, Trisomy 13 & 18 and Spina Bifida. Down Syndrome babies have mental retardation of varying degrees and some physical changes in their face and body. They often have other birth defects, such as congenital heart disease. The risk of having a baby with Down Syndrome increases with age. Trisomy 13 &18 are rare genetic disorders that involve many serious mental and physical disabilities. There is no cure and it is usually fatal. Spina Bifida is an opening in the baby's spine or incomplete development of the brain. This occurs in 1 or 2 out of every 1,000 births. The effect of spina bifida depends on where the defect is located in the spine. Effects could be mild or severe, including paralysis. If your Quad Screen is positive, it means that you are at higher risk for one of these problems but we cannot be sure until further testing is done. Remember, in most cases the baby is healthy, even if the screen results are abnormal.

LEVEL II ULTRASOUND:

This ultrasound is only offered to women over 35, women with a positive Quad Screen, and to women with other specific risk factors. A level II ultrasound can provide more detailed information than the traditional level I ultrasound offered to all women. Insurance usually covers this test. It is done between 18-20 weeks gestation. A level II ultrasound cannot rule out diseases such as Down Syndrome or Spina Bifida, but it can help us to better understand your risks.

AMNIOCENTESIS:

Also known as "amnio," this test is only offered to women over 35, women with positive Quad Screen, and to women with other specific risk factors. Insurance usually covers this test. An amniocentesis involves taking a sample of amniotic fluid under ultrasound guidance and analyzing it. There are some risks to the procedure and it is usually done between 16-17 weeks gestation. This procedure can detect Down Syndrome, Spina Bifida, and Trisomy 13 & 18. It can be used to test for other diseases but this testing is ONLY done in women who are known to be at high risk or carriers of these diseases. Talk with your midwife or doctor if you think you are at high risk for other diseases.

TAY SACH, GAUCHER'S AND CANAVAN'S DISEASE:

Eastern and Central European Jew (Ashkenazi Jews) and French-Canadians are most at risk for these diseases. Insurance may cover. If not, the testing can cost up to $1,000. The testing is a blood draw from the mother, and can be done at any time, including before getting pregnant. If both parents carry a gene for one of these diseases, the fetus is a risk for the disease. Please refer to your book for details on these specific diseases.