Plano Women's Healthcare

Optional Tests Offered Before and During Pregnancy

Alpha-Fetoprotein Test (AFP) and Quad Screen

These are screening tests that can assess your baby's risk of having such birth defects as Spina Bifida, Anecephaly and Downs Syndrome. As with all *screening* tests, the results are not 100%. Not every abnormal result will mean that your baby has a birth defect. Likewise, not every normal result guarantees that the baby is defect free. In the cases where an abnormal result is reported, diagnostic tests such as a level two sonogram or an amniocentesis should be preformed.

Spina Bifida is a neural tube defect that can result in the baby's brain and or spinal cord to remain exposed (not covered with skin). Symptoms of this may include:
Leg paralysis
Lack of bladder and bowel control
Scoliosis (curvature of the spine)
Hydrocephalus (increased fluid on the brain)
Mental Retardation
Death

Anecephaly is another form of neural tube defect. With this the baby's brain and head do not develop normally. As this condition is not compatible with life, babies with anencephaly are either stillborn or die shortly after birth.

Down Syndrome is a genetic disorder caused by trisomy 21. Normally, there are 23 pairs of chromosomes. In the case of Down syndrome, there is an extra copy of chromosome #21. This causes mental retardation to varying degrees, heart defects, and abnormal facial features such as flat face and low set ears. The risk for having a baby with Down syndrome increases with maternal age. The risk at age 35 in 1 in 378.

Both the AFP and the Quad Screen are done from a small amount of the mother's blood drawn at 15- 18 weeks gestation. Usually results are available within 2 weeks. As this is an optional test, it may not be covered by all insurances. Please check with your insurance company to verify coverage. Out of pocket costs could reach $300.

It is your choice whether to be tested. Some families find the tests reassuring while others would rather not have the information. The results can help some women make decisions about their options.

Cystic Fibrosis Carrier Testing

Cystic Fibrosis is a genetic disorder that is usually diagnosed during the first few years of life. Both parents must be carriers for the baby to develop CF. While this disease does not affect intelligence or appearance, its affects on the digestive system and the lungs are serious. Those children with CF must undergo daily respiratory therapy treatments as well as taking daily doses of medicine to treat the digestive system.

Genetic testing is done from a small sample of blood from the mother and father. While there are some mutations that the current test cannot find, the likelihood that you are a carrier when the results were reported normal is small. The cost of testing is covered by some insurance. Please check with your insurance carrier before deciding to proceed with testing.

Ethnicity /Race	Chance of both parents being carriers
European Caucasian, Ashkenazi Jewish	1 in 841
Hispanic American	1 in 2,116
African American	1 in 4,225
Asian American	1 in 8,100

For more information:

Cystic Fibrosis Foundation	WWW.CFF.ORG
National Society of Genetic Counselors	WWW.NSGC.ORG
Genetic Alliance	WWW.Geneticalliance.ORG

Cord Blood Banking

Cord blood banking is the preservation of the blood from the baby's umbilical cord at birth. This blood contains the building blocks of all cells called stem cells. These stem cells divide to create the white blood cells of the immune system, red blood cells that carry oxygen to tissues and vital organs, and platelets, which are responsible for clotting. Collection of cord blood is time restricted. It can only be retrieved immediately after birth so it is important to have preparations ready for its collection prior to delivery.

The stem cells are used similarly to how bone marrow is used presently. The transplantation of stem cells is being used to treat a wide range of serious diseases including cancer, leukemia, lymphoma, some forms of anemia, sickle cell and other immune deficiencies. So should your baby become seriously ill, the materials needed for treatment are readily available because they have been cryogenically frozen to preserve their inherent value. Because stem cells are collected from the infant before it has been exposed to any disease, they are a perfect, uncompromised match for your baby so the risk of rejection is null. In addition, banked cells have a 1 in 4 chance of being an exact match for the baby's siblings. This could be a major point for ethnic minorities who, because of low donation rates to the National Marrow Donor Program, could have difficulty locating suitable transplant material.

There are many cord blood banks from which to choose from. The web addresses for some of the larger companies have been provided. If you are considering banking your infant's cord blood, please take the time to review your choices.

www.viacord.com
www.cordblood.com
www.cryo-cell.com
www.securacell.com
www.lifecd.com

Thyroid Stimulating Hormone

A 3- year study published in The New England Journal of Medicine found that the children of women who were not treated for Hypothyroidism in pregnancy averaged 7 points lower on IQ tests and that nearly 1 in 5 (19%) had scores of 85 or less. High serum TSH concentrations were to blame. While thyroid disease is relatively easy to treat early diagnosis is the key.

So, to avoid the unnecessary complications brought on by undiagnosed hypothyroidism, we have chosen to offer a routine TSH screening to all obstetrical patients.

Your insurance company may not cover this test due to plan limitations or lack of medical necessity, in which case, you will be financially responsible for the cost of the test.

Hemoglobinopathies

This family of genetic disorders affects the body's ability to transfer oxygen to the cells. There are several different types of hemoglobinopathies the most common include Sickle cell and Thalassemia. While these anemias can occur among all ethnic and racial groups, studies have shown an increase in occurrence among those of African, Southeast Asian, Mediterranean, and Middle Eastern decent. The average life expectancy of patients with Sickle Cell Anemia is decreased by 25-30 years. During which time symptoms could include, painful crisis, stokes, splenic and renal dysfunction, and bone and joint disease. Parents who are both carriers have a 25% probability of having a child with Sickle Cell. Among African Americans that translates to 1 in every 150 couples.

Testing for this carrier status includes collecting a small amount of blood from the mother. If results are positive for carrier state then testing of the father is recommended. Once both parents have been identified as carriers then an amniocentesis can be performed to evaluate the state of the fetus.

For more information on Sickle Cell and Thalassemia please refer to the many valuable web based informational sites by searching for the key word 'Hemoglobinopathies'.

Tay Sachs Disease

TSD is a fatal genetic disorder in children that causes progressive degradation of the central nervous system. The disease usually begins to show it effects when the infant is several months old. The most common symptom noticed includes a slow regression in ability. The child will loose skills such as crawling, coordination, breathing and swallowing. Even with the best of care, all children with TSD die in early childhood.

Studies show that 1 in every 27 Jews in the United States is a TSD gene carrier. In addition, there is an increased incidence in French Canadians and the Cajun community of Louisiana. When both parents are carriers of the inactive gene, they have a 1 in 4 chance that their child will have Tay Sachs.

A simple blood test can distinguish carriers from non-carriers. The best advise for child bearing aged women is to be tested before pregnancy. That way, if a couple is found to be at risk, they can review their options and make the necessary decisions about planning and protecting their families.

For more information please contact:

National Foundation for Jewish Genetic Diseases, Inc. www.nfjgd.org
National Tay-Sachs & Allied Diseases Association, Inc. www.ntsad.org

Canavan Disease

Canavan Disease is an inherited enzyme deficiency and is characterized by developmental delays in infancy. While most infants appear normal early in life, they soon display delays in motor skills and lack of head control. Life expectancy for those with Canavan Disease varies.

It has been shown that there is an increase in frequency of this genetic deficiency in people of Ashkenazi Jewish heritage. Carrier rates are estimated at 1 in 40. Because Canavan Disease is a recessive disorder, each pregnancy in which both parents are carriers can result in a 25% chance of producing a child that is affected, a 50% chance of producing an unaffected child that is a carrier, and a 25% chance in producing a child that is unaffected and not a carrier.

Carrier status is established in the parents through a simple blood test. Fetal status is determined through amniotic fluid collection during weeks 16-18 of the pregnancy.

For resources and more information please contact:

Canavan Foundation
Toll free 1-877-4-Canavan
www.canavanfoundation.org

Vitamin D Level

The maternal vitamin D level has an effect on fetal acquisition of bone mineral in utero. Vitamin D deficiency can cause growth retardation and skeletal deformities and may increase the risk of hip fracture in later life. Fetal stores of vitamin D depend entirely on maternal supply. Most prenatal vitamins contain only 400IU of vitamin D daily. This amount may not be sufficient for both mother and developing fetus.
It is important to include assessment of 25-hydroxyvitamin D to determine which patients require supplementation to prevent detrimental fetal effects.