

Prince William OBGYN

MATERNAL SERUM SCREENING

Neural tube defects are serious birth defects that include the brain and spinal cord and may result in paralysis of limbs, mental retardation or death. They occur in approximately 1/1000 births in those families without a family history of such abnormalities. There is no warning during pregnancy that this abnormality exists. There is a blood test available (drawn from the mother) which will detect the presence of neural tube defects 90% of the time. This test is called the alpha fetoprotein test (AFP).

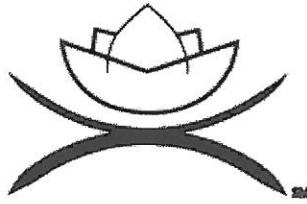
Also, the addition of measuring three other hormones will increase the detection of Down's Syndrome (a genetic form of mental retardation). This test is known as the quadruple test.

Both the AFP and the quadruple test are best done during 15-20 weeks of pregnancy using blood drawn from the mother. If abnormal, this blood test must be followed by a very careful evaluation which may include a repeat blood test for confirmation, examination of the baby in the uterus by ultrasound, and, possibly, taking some of the fluid from around the baby for testing (amniocentesis). The reason for such careful testing is that only 10% of women with positive tests will have an abnormal baby. Most of the positive tests are due to other causes, such as an error in estimating the duration of pregnancy, the presence of more than one baby, or the presence of an abnormality other than a neural tube defect. The only definite way to determine if there is a chromosomal abnormality is by amniocentesis.

The tests are not perfect. The chance that the blood test will not detect a neural tube defect is about 10%, and the chance of not detecting Down's Syndrome is approximately 20%. Stated another way, this means that these tests will detect 90% of neural tube defects and 80% of Down's Syndrome.

It is recommended that those women under 35 who wish to be evaluated for Down's Syndrome have the quadruple test. Women age of 35 or older are encouraged to have a more advanced genetic test such as chorionic villus sampling (CVS) or amniocentesis, because their risk of having a chromosomal abnormality, such as Down's Syndrome, is increased. Women who received or will receive amniocentesis will have the AFP testing done at the time of the amniocentesis.

You may obtain either the Alpha Fetoprotein (AFP) or quadruple test by asking your doctor, who will arrange to have your blood drawn. The results of the tests are usually available in about one week. These tests are usually covered by insurance, but you may want to check with your company prior to having it drawn.



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PRENATAL SCREENING QUESTIONNAIRE

PATIENT NAME: _____ DATE: _____

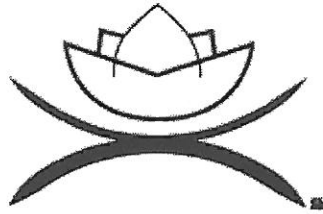
- 1.) Will you be age 35 or older when the baby is due? Yes _____ No _____
- 2.) Have you, the baby's father, or any blood relatives had:
 - Down's Syndrome (mongolism)? Yes _____ No _____
 - Spina Bifida (open spine), or Anencephaly? Yes _____ No _____
 - Hydrocephaly (water on the brain)? Yes _____ No _____
 - Hemophilia (bleeding disorder)? Yes _____ No _____
 - Muscular dystrophy? Yes _____ No _____
 - Cystic fibrosis? Yes _____ No _____
- 3.) Have you, the baby's father or any blood relatives had a child born dead or alive with a birth defect not listed in question 2? Yes _____ No _____
If so, please describe _____
- 4.) Do you, the baby's father or any blood relatives have any inherited genetic or chromosomal disease or disorder not listed above? Yes _____ No _____
- 5.) Do you, the baby's father have any blood relatives with mental retardation? Yes _____ No _____
- 6.) Have you had three or more spontaneous pregnancy losses? Yes _____ No _____
- 7.) Do you or the baby's father have any blood relatives descended from Jewish people who lived in Europe (Ashkenazi Jews)? Yes _____ No _____
- 8.) Have you, the baby's father or any close relatives been screened for sickle cell trait or disease and found to be positive? Yes _____ No _____
- 9.) Do you have any known blood antibodies? Yes _____ No _____
- 10.) During this pregnancy, have you used:
 - Alcohol? Yes _____ No _____
 - Tobacco? Yes _____ No _____
 - Drugs? Yes _____ No _____
 - Medications? Yes _____ No _____
 - Toxic Substances? Yes _____ No _____
- 11.) Do you have any cats? Yes _____ No _____

I have read and understand the information sheet on the First Trimester Prenatal Screening and Alpha Fetoprotein Testing. Please circle all that apply:

- A. I wish to have the First Trimester Screening (nuchal translucency, PAPP-A And freeBeta-hCG)
- B. I wish to have the AFP Tetra screening
- C. I wish to have testing with CVS or amniocentesis
- D. I do not wish to have any testing or screening
- E. I am not eligible for testing because I am more than 20 weeks pregnant.
- F. I wish further information/counseling on genetic testing/screening

Patient's Signature: _____

Doctor's Initials _____



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NEWBORN SCREEN

Please answer the following questions so that the nursery may prepare for the arrival of your newborn.

- 1) Have you ever had a baby who was infected with GBS during or after delivery? ()NO ()YES
- 2) Have you ever had a newborn or infant stop breathing in the first year of life that required emergency room support with hospitalization to recover? ()NO ()YES- if yes, was the infant put on a ventilator /respirator (breathing machine) to recover? ()NO ()YES
- 3) Have you ever had a newborn or infant in the first year of life suspected to have Sudden Infant Death Syndrome or SIDS? ()NO ()YES
- 4) Have you ever had a newborn or an infant in the first year of life sent home with a cardiac/respiratory monitor or oxygen? ()NO ()YES
- 5) Have you ever had a newborn that needed to stay in the hospital, after you were discharged following delivery? ()NO ()YES Why? _____

NAME _____ Date _____

Physician Reviewed _____



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Cystic Fibrosis Carrier Testing

What is Cystic Fibrosis? Cystic fibrosis is a genetic disease that affects about 1 in 3300 people in the United States. Symptoms of the disease range from mild to severe. Most often, people with CF have chronic lung problems that will continue to worsen over time. CF may also affect digestion resulting in chronic diarrhea and poor weight gain and growth. There are some treatments available for the lung and digestive problems, but most people with CF have a shortened lifespan. Most people do not reach adulthood with CF. Men are almost always infertile. CF does NOT affect intelligence.

What is a CF carrier? Each person has two copies of a gene, one inherited from each parent. If a person has one normal CF gene and one abnormal CF gene, that person is a carrier of the disease and, therefore, does not have symptoms of the disease, but may be able to transmit this risk to offspring, including the disease. This would happen if both parents were carriers of the same abnormal gene. In this scenario, the risk of disease would be 25% (1 out of 4). The risk of carrier state would be 50% (2 out of 4) and the risk of complete normal genes, would be 25%.

Can anyone be a CF carrier? Yes. Your ethnic background can determine your carrier state risk. CF is more common in people of Caucasian and Ashkenazi Jewish descent. CF is less common in Hispanic, African American, Native American, or Asian backgrounds. If someone in your family has CF, then your risk of being a carrier is increased above what your risk is based on ethnic background.

What is the purpose of carrier testing? The purpose of testing is to see if a couple is at increased risk for giving birth to a child who will have CF. Carrier-testing is a laboratory test on blood used to show if a couple is at high risk. Additional testing on the developing baby may then be recommended.

If my test is negative, could I still be a carrier? The answer to this is "yes." There are some mutations of the CF gene that the current test cannot find. Currently the test includes a minimum of 25 specific CF gene mutations, most of which are common in the higher risk ethnic groups. Like most medical tests, there are limitations because not all the CF mutations are known. However, these mutations are rare.

INFORMED CONSENT:

1. I understand that the decision to be tested for CF carrier status is completely mine.
2. I understand that the test does not detect all CF carriers.
3. I understand that if I am a carrier, testing the baby's father will help me learn more about the chance that my baby could have CF.
4. I understand that if one parent is a carrier, and the other is not, it is still possible that the baby will have CF, but the chance of this is very small.
5. I understand that if both parents are carriers, additional testing can be done in order to know whether or not the baby will have CF.
6. I understand that if the baby has inherited a changed CF gene from each parent, that the baby will have the disease Cystic Fibrosis, and further genetic counseling and recommendations from a perinatal center will be offered.

I have read and understand the information presented to me regarding CF testing.

_____ I do not want CF carrier testing.

_____ I want CF carrier testing.

Patient: _____ DOB: _____ Signed: _____ Date: _____

Witness: _____ Date: _____

(Please note that insurance coverage for this test is dependent upon your own individual insurance policy).