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PATIENT INFORMATION FOR GENETIC SCREENING

genetic screening

Many options are now available for prenatal screening for chromosomal defects. Down syndrome (also called Trisomy 21) is the most common cause of severe learning disabilities in children. There is no way to prevent Down syndrome, but there are tests to indicate if the fetus is at increased risk of having a chromosomal abnormality. A chromosomal abnormality may affect any pregnancy but some pregnancies are at higher risk than others. Risk factors include mothers who are older, parents with a previous baby with a chromosomal abnormality, and parents with a chromosomal abnormality. Prenatal screening tests are voluntary and about 60% of women in the United States undergo screening.

Screening Tests help determine the risk of having an affected fetus but are not diagnostic and therefore would require further testing to make a diagnosis. Screening tests consist of blood tests in the first trimester, second trimester, and or ultrasound or a combination of these. **Diagnostic tests** consist of chorionic villous sampling (CVS) or amniocentesis. The most common screening tests are:

1. **Serum Integrated Test**: Consists of first trimester blood tests and second trimester blood tests.
2. **Combined (BUN) Test**: Consists of first trimester blood tests and a special early ultrasound measurement.
3. **Quadruple Screening**: Consists of second trimester blood tests.
4. **Fully Integrated Screen**: Combines tests 2 and 3

The BUN test is performed from 11 weeks, 0 days to 13 weeks, 6 days of gestation; the optimal time is 11 weeks. This consists of two blood tests and a highly specialized ultrasound measurement looking at the nuchal fold on the back of the fetus's neck. The results are known by 14 weeks gestation allowing for CVS to make definitive diagnosis early in the pregnancy. Disadvantages include concerns regarding variability in operator expertise and quality of equipment within the studies used to evaluate this test and occasionally the inability to view the area required. This test also does not screen for neural tube defects. An additional test for this can be done at 15-19 weeks.

A Serum Integrated Test combines serum markers measured in the first trimester and the Quadruple test measured in the second trimester to determine a single estimate of risk for Downs's syndrome, T18 pregnancy and risk for neural tube defects. Some advantages to this test are in improved detection rate while only requiring two blood draws. The main disadvantage being that the results are not known until the second trimester.

The Quadruple Test measures serum markers in the second trimester to detect Down syndrome, T18 pregnancies and neural tube defects. Some advantages to the test are detection of the main defects of pregnancy, requiring only a single blood draw, and all tests are done in the second trimester. The main disadvantage is a slightly lower detection rate and results are not available until the second trimester.

A **positive screening test** refers to a result showing a woman's risk of having an abnormality.

For the BUN test, the risk cutoff is 1 in 220 for Down syndrome, and 1 in 100 for trisomy 18. *No risk analysis for neural tube defects is available with this test, but is available with a separate second trimester blood test.*

For the Serum Integrated Test, a positive test result confers a risk of more than 1 in 270 for Down syndrome and 1 in 100 for trisomy 18, and a risk of approximately 1 in 145 for NTDs depending on ethnicity and possible diabetes.

In the Quadruple Screen, a positive result confers a risk of more than 1 in 190 and of a trisomy 18 risk more than 1 in 100 in the mid-trimester, and for neural tube defects a risk more than 1 in 145, again depending on ethnicity and possible diabetes.

Chorionic villous sampling (CVS) is a test that can be used to examine the baby's chromosomes during the first trimester of pregnancy. The test involves using a needle or catheter which is inserted through the abdomen or the cervix, to remove a tissue sample from the placenta which is then examined for chromosomal abnormalities. There is a procedure-related risk of miscarriage of about 1 in 100 women who have CVS.

Amniocentesis is a test used to examine the baby's chromosomes after 14 weeks gestation. The test involves using a needle inserted through the abdomen to extract some amniotic fluid from the uterus. The fetal cells in the fluid are examined for chromosomal problems. There is a procedure-related risk of miscarriage of about 1 in 200-300 women who have an amniocentesis.

CYSTIC FIBROSIS

Cystic fibrosis is a lifelong genetic disorder causing problems with digestion and breathing. Both parents must be carriers for the baby to develop cystic fibrosis. Some individuals with cystic fibrosis have a milder form, while others have more severe symptoms. Carriers will generally have no symptoms at all. In general, people with cystic fibrosis have a shorter life span and may die in childhood while others live into their 40s or longer. There is no cure for CF. CF cannot be treated before delivery. The purpose of having this information about your baby is to prepare yourself to care for the child with health-care needs or so you can terminate the pregnancy.

COULD I BE A CARRIER of CYSTIC FIBROSIS?

About 3% (1 out of every 30) of the white population are carriers, while other racial groups have less risk of being a carrier:

Ethnicity/Race	Chance of 1	Chance of both	Chance of baby being affected
European Caucasian/Ashkenazi Jewish	1 in 29	1 in 841	1 in 3,364
Hispanic American	1 in 46	1 in 2,116	1 in 8,464
African American	1 in 65	1 in 4,225	1 in 16,900
Asian American	1 in 90	1 in 8,100	1 in 32,400

There are some mutations of the cystic fibrosis gene the test cannot find. If you have this mutation you would be told your test result is normal, and you would still be a carrier. If the test shows you are a carrier, the next step is to test the baby's father as both parents must be positive for the baby to have cystic fibrosis. If both parents are carriers and you decide to test the baby this could be done about the 11th-14th week of pregnancy using chorionic villous sampling which removes a small piece of placenta or about the 16th week of pregnancy by amniocentesis. After learning about cystic fibrosis carrier testing, some people decide to have the test while others do not. The cost of testing is covered by some insurances and not by others.

REASONS TO BE TESTED:

- If you are at increased risk of being a carrier or if there is a family history of CF
- If you and the baby's father were both found to be CF carriers and you would consider an amniocentesis or chorionic villous sampling to help you decide to continue the pregnancy or prepare for the birth of a baby with CF

REASONS NOT TO BE TESTED:

- If the chance of being a CF carrier seems low
- Even if you were both found to be carriers, if you and the baby's father would never consider having an amniocentesis or chorionic villous sampling to help you decide about terminating the pregnancy or preparing for the birth of a baby with CF.

The choice to pursue testing is highly individual and personal. There are a number of reasons why a woman would choose not to have genetic screening, such as not wanting to expose the pregnancy to the potential risks related to more invasive tests needed to confirm the diagnosis if screening is positive and she is willing to wait until the baby's birth to find out about one of these diagnoses or she would not terminate the pregnancy if the fetus had an abnormality. However, even if she would not terminate the pregnancy, she may wish testing in order to better prepare for the birth of a baby who may have special needs.

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GENETIC SCREENING CONSENT FORM

genetic screening

I have received and reviewed the patient information regarding genetic screening and cystic fibrosis carrier screening. After discussion with my health care provider, I wish to have the following genetic testing:

_____ **Serum Integrated Test**
_____ **BUN Test**
_____ **quadruple Test**
_____ **Cystic Fibrosis Carrier Screening**

_____ _____
(Patient Signature) Date

_____ **I wish to decline genetic screening**

_____ _____
(Patient Signature) Date

_____ _____
(Physician Signature) Date

SCAN Under Consent*