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First Trimester Screening

Screening for Down Syndrome and Trisomy 18

When a pregnant woman sees her doctor in the first trimester of pregnancy she may be offered a screening test for Down Syndrome and Trisomy 18. This recent advancement in prenatal care is called first trimester screening. First trimester screening is designed to identify women who may be at increased risk of having a baby with Down Syndrome or Trisomy 18. It is important to remember that this is a screening test and that even when it is "screen positive," most often the baby is healthy. It is also important to remember that not every baby with Down Syndrome or Trisomy 18 will be identified with this screen.

First trimester screening involves a specific ultrasound measurement called "nuchal translucency" and a maternal blood draw. First trimester screening assesses the chance of having a baby with Down Syndrome and Trisomy 18. It is typically drawn between 11 and 13 weeks 6 days gestation. Results take approximately 6-7 days.

What are Chromosome Abnormalities?

Humans typically have 46 chromosomes in every cell. These chromosomes carry genes, which are the blueprints for human growth and development. If a child is born with more or less than 46 chromosomes in each cell, he or she will have problems with mental and physical development. A few examples of chromosome abnormalities are Down syndrome (Trisomy 21) and Trisomy 18. These two chromosome conditions are screened for with first trimester screening.

Down Syndrome is a chromosome problem that occurs when a baby inherits an extra chromosome #21 at conception. This extra chromosome causes the features of Down Syndrome, which can include heart defects, mental retardation and other physical disabilities. Down Syndrome occurs in 1:800 pregnancies in the general population. The risk of having a baby with Down Syndrome increases with increasing maternal age. However, 80% of babies with Down Syndrome are born to women less than 35 years of age. Most couples who have a child with Down Syndrome have no family history of the condition.

Trisomy 18 is a chromosome problem that occurs when the baby inherits an extra chromosome #18 at conception. It is medically more severe than Down Syndrome and babies with Trisomy 18 typically will be stillborn or will not live past one year of age. Babies that do survive will have mental retardation and can have a variety of other severe birth defects.

What is First Trimester Screening?

In first trimester screening two chemicals are measured from a pregnant women's blood: free beta-human chorionic gonadotropin (free B-hCG) and pregnancy-associated plasma protein A (PAPP-A). In addition, an ultrasound measurement of the baby's neck is performed to determine the amount of fluid accumulation behind the neck of the baby. This measurement is called "nuchal translucency." The amounts and patterns of the chemicals are used, along with the nuchal translucency measurement and the mother's age, to assess the likelihood that a baby

would have Down Syndrome or Trisomy 18. A positive screen means that the calculated chance of having a baby with Down Syndrome is greater than or equal to the woman's age based risk. It is important to note that the levels of these chemicals are not usually affected by anything mom may or may not have done during the pregnancy.

How Accurate is First Trimester Screening?

In all women, first trimester screening identifies approximately 90% of babies who have Down Syndrome and about 97% of babies who have Trisomy 18. **Again, it is important to remember that most of the time, even when the screen is positive, the baby does not have a problem.** It is also important to remember that this is a screening tool and it will not detect every baby with Down Syndrome or Trisomy 18.

What does a Positive Result Mean?

There can be several different explanations for a positive result. A positive result does not mean Down Syndrome or Trisomy 18 has been diagnosed. You should talk to your doctor or genetic counselor in order to get more information about your particular situation.

My Result was Positive, Now What?

Again, when the first trimester screen is positive, it does not mean that your baby has been diagnosed with a problem. It tells us that we should offer you additional prenatal tests, such as chorionic villus sampling (CVS) or amniocentesis, to give you more information about your pregnancy. CVS is the removal of a small sample of tissue from the placenta, which allows us to perform chromosome analysis. Amniocentesis is a procedure that involves withdrawing a small sample of amniotic fluid; this allows us to look at the baby's chromosomes and to test the AP levels. However, amniocentesis cannot tell us everything about the baby and is associated with a risk of miscarriage (1:200). A discussion with your doctor or genetic counselor can help you understand these procedures and the benefits, risks, and limitations of these options.

It is your decision whether or not to have the test. Some women find having the test to be reassuring. Some face an anxious time between a “positive” screen and the follow up testing. Others would rather not have the information. The results can help some women make decisions about their options.

What Other Tests Should I Have?

A blood test should be done between 15-21 weeks is recommended to screen for 98% of open spine and skull defects, and about 60% of abdominal wall defects. These birth defects are not screened for with first trimester screening and usually occur without a family history. Ultrasound examination in the second trimester is another way to screen for these types of defects.

Should I have the Quad Screen if I have the First Trimester Screening?

No. A second trimester chromosomal screen is not recommended. This is because any second trimester screening is unlikely to improve the detection rate and there is greater chance of a false positive result.