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| SCWHCA_1 | **Southern Connecticut Women’s Health Care Associates****247 Broad Street, Milford CT 06460****P. (203) 783-0543 – F. (203) 874-5728** | Miriam Sivkin, M.D. FACOGMary Murray, M.D. FACOGErika Schaeffer, M.D. |

**What Is the First-Trimester Screening?**

First trimester screening is a prenatal test that offers early information about a baby's risk of certain chromosomal conditions — Down syndrome (trisomy 21) and extra sequences of chromosome 18 (trisomy 18).

**First trimester screening, also called the first trimester combined test, has two steps:**

* A blood test to measure levels of two pregnancy-specific substances in the mother's blood — pregnancy-associated plasma protein-A and human chorionic gonadotropin (HCG)
* An ultrasound exam to measure the size of the clear space in the tissue at the back of the baby's neck (nuchal translucency)

Typically, first trimester screening is done between weeks 11 and 14 of pregnancy. Using your age and the results of the blood test and the ultrasound, your health care provider can gauge your risk of carrying a baby with Down syndrome or trisomy 18. If results show that your risk level is moderate or high, you might choose to follow first trimester screening with another test that's more definitive.

## **Why it's done**

First trimester screening is done to evaluate your risk of carrying a baby with Down syndrome. The test also provides information about the risk of trisomy 18. Down syndrome causes lifelong impairments in mental and social development, as well as various physical concerns. Trisomy 18 causes more severe delays and is often fatal by age 1.

First trimester screening doesn't evaluate the risk of neural tube defects, such as spina bifida.

Because first trimester screening can be done earlier than most other prenatal screening tests, you'll have the results early in your pregnancy. This will give you more time to make decisions about further diagnostic tests, the course of the pregnancy, medical treatment and management during and after delivery. If your baby is diagnosed with Down syndrome, you'll also have more time to prepare for the possibility of caring for a child who has special needs.

Other screening tests can be done later in pregnancy. An example is the quad screen, a blood test that's typically done between weeks 15 and 20 of pregnancy. The quad screen can evaluate your risk of carrying a baby with Down syndrome or trisomy 18, as well as neural tube defects, such as spina bifida. Some health care providers choose to combine the results of first trimester screening with the quad screen. This is called integrated screening. This can improve the detection rate of Down syndrome.

Remember, first trimester screening is optional. Test results only indicate whether you have an increased risk of carrying a baby with Down syndrome or trisomy 18, not whether your baby actually has one of these conditions.

Before the screening, think about what the results will mean to you. Consider whether the screening will be worth any anxiety it might cause, or whether you'll manage your pregnancy differently depending on the results. You might also consider what level of risk would be enough for you to choose a more invasive follow-up test.

## **Risks**

First trimester screening is a routine prenatal screening test. The screening poses no risk of miscarriage or other pregnancy complications.

## **How you prepare**

You don't need to do anything special to prepare for first trimester screening. You can eat and drink normally before both the blood test and the ultrasound exam.

## **What you can expect**

First trimester screening includes a blood draw and an ultrasound exam. During the blood test, a member of your health care team takes a sample of blood by inserting a needle into a vein in your arm. The blood sample is sent to a lab for analysis. You can return to your usual activities immediately.

For the ultrasound exam, you'll lie on your back on an exam table. Your health care provider or an ultrasound technician will place a transducer — a small plastic device that sends and receives sound waves — over your abdomen. The reflected sound waves will be digitally converted into images on a monitor. Your health care provider or technician will use these images to measure the size of the clear space in the tissue at the back of your baby's neck.

The ultrasound doesn't hurt, and you can return to your usual activities immediately.

## **Results**

Your health care provider will use your age and the results of the blood test and ultrasound exam to gauge your risk of carrying a baby with Down syndrome or trisomy 18. Other factors — such as a prior Down syndrome pregnancy — also might affect your risk.

First trimester screening results are given as positive or negative and also as a probability, such as a 1 in 250 risk of carrying a baby with Down syndrome. First trimester screening correctly identifies about 85 percent of women who are carrying a baby with Down syndrome. About 5 percent of women have a false-positive result, meaning that the test result is positive but the baby doesn't actually have Down syndrome.

When you consider your test results, remember that first trimester screening only indicates your overall risk of carrying a baby with Down syndrome or trisomy 18. A low-risk result doesn't guarantee that your baby won't have one of these conditions. Likewise, a high-risk result doesn't guarantee that your baby will be born with one of these conditions.

After a positive test result, your health care provider and genetics professional will discuss your options, including additional testing. For example:

* **Prenatal cell-free DNA (cfDNA) screening.** This is a sophisticated blood test that examines fetal DNA in the maternal bloodstream to determine whether your baby is at risk of Down syndrome, extra sequences of chromosome 13 (trisomy 13) or extra sequences of chromosome 18 (trisomy 18). Some forms of cfDNA screening also screen for other chromosome problems and also provide information about fetal gender. A normal result might eliminate the need for a more-invasive prenatal diagnostic test.
* **Chorionic villus sampling (CVS).** CVS can be used to diagnose chromosomal conditions, such as Down syndrome. During CVS, which is usually done during the first trimester, a sample of tissue from the placenta is removed for testing. CVS poses a small risk of miscarriage.
* **Amniocentesis.** Amniocentesis can be used to diagnose both chromosomal conditions, such as Down syndrome, and neural tube defects, such as spina bifida. During amniocentesis, which is usually done during the second trimester, a sample of amniotic fluid is removed from the uterus for testing. Like CVS, amniocentesis poses a small risk of miscarriage.

Your health care provider or a genetic counselor will help you understand your test results and what the results mean for your pregnancy.