BRIGHAM AND WOMEN'S HOSPITAL

The First Trimester Test

Prenatal Screening for Down Syndrome

What is the First Trimester Test?

The First Trimester Test is performed between 11 and 13 completed weeks of pregnancy to screen for Down syndrome. It combines information from an ultrasound examination of your baby with maternal blood analysis. It is suitable for women of all ages. It is a screening test and cannot determine definitely whether or not a baby has Down syndrome. A screening test identifies those women who have an increased risk of Down syndrome pregnancy so that they can be offered a diagnostic test (such as chorionic villus sampling). The diagnostic test identifies women who actually have an affected pregnancy.

The information below answers some of the common questions women ask about the screening test. We hope you find it helpful. You are welcome to discuss the test with your health care provider before you decide whether you would like to be screened. If you have any further questions, staff at Women & Infants are available to talk with you at 401-453-7650.

Is the First Trimester Test right for me?

The First Trimester Test should be chosen if you want prenatal screening and diagnosis as early as possible.

However, you should be aware that another screening test, called the Integrated Test, provides the same level of detection with about 75% fewer women having to consider invasive prenatal diagnostic procedures. The Integrated Test is done in two stages and combines parts of the First Trimester Test with a blood test done about four weeks later, to provide a single screening result in the early second trimester. The Integrated Test also provides screening for neural tube defects, while the First Trimester Test does not.

What is Down syndrome?

Down syndrome is caused by the presence of an extra chromosome number 21 in the cells of the developing baby. In an unscreened population, about 1 in every 700 babies is born with Down syndrome. Usually it is not inherited and so a baby can be affected even if there is no history of Down syndrome in the family. Although Down syndrome occurs more frequently as mothers get older, about 70% of babies with Down syndrome are born to women who are younger than 35 years old.

Down syndrome is always associated with intellectual disability, and is often associated with physical problems such as heart defects and difficulties with sight and hearing. It is not possible to assess the degree of handicap before the baby is born. About 9 out of 10 babies with Down syndrome will survive their first year, and nearly half of these will reach 60 years of age.

What does the First Trimester Test involve?

A sample of your blood is taken between 11 and 13 weeks of pregnancy. At the same time, an ultrasound scan is performed.

The substances in your blood that are markers of Down syndrome are:

(i) pregnancy associated plasma protein A (PAPP-A); and
(ii) human chorionic gonadotropin (hCG).

The specific ultrasound marker of Down syndrome is:
(iii) nuchal translucency (NT) thickness.

In pregnancies with Down syndrome, PAPP-A tends to be low, and NT and hCG tend to be raised.

The values of these markers are used together with your age to estimate the risk of having a pregnancy affected with Down syndrome.

What is a "risk"?

A risk is the chance of an event occurring. For example, a risk of Down syndrome of 1 in 100 means that if 100 women have this risk, we expect that 1 of these women will have a baby with Down syndrome and that 99 will not. This is the same as saying that the baby has a 1% chance of having Down syndrome and a 99% chance that it does not.

When will the results be available?

The results of the test are usually ready within 3 working days of the blood sample being taken. Results are sent to your health care provider.

Your screening result is either *screen positive* or *screen negative*. *Screen positive* results are telephoned and/or faxed to your health care provider.

What does a *screen positive* result for Down syndrome mean?

A *screen positive* result means that you are in a high risk group for having a baby with Down syndrome. If you are in this group, you will be offered a diagnostic test.

The result is *screen positive* if the risk of Down syndrome in the first trimester is 1 in 230 or greater. About 1 in every 20 women screened will be in this group.

Most women with *screen positive* results do not have a pregnancy with Down syndrome. For example, of about 50 women with *screen positive* results for Down syndrome, only 1 would have an affected pregnancy.

What does a *screen negative* result mean?

If the risk of Down syndrome based on the first trimester test is lower than 1 in 230, then the result is called *screen negative* and a diagnostic test is usually not offered.

Although a *screen negative* result means that your risk of having a baby with Down syndrome is not high, **a *screen negative* result cannot rule out the possibility of a pregnancy with Down syndrome.**

Does the First Trimester Test detect all pregnancies with Down syndrome?

No. About 8 or 9 out of 10 cases of Down syndrome are detected (classified as *screen positive*). This means that 1 or 2 out of 10 pregnancies with Down syndrome are missed (classified as screen negative).

Why do women with *screen negative* results occasionally have babies with Down syndrome?

It is uncommon for a woman to have a baby with Down syndrome and it is even more uncommon for a woman with a *screen negative* result, but it does sometimes happen.

This is because the screening test cannot completely distinguish affected from unaffected pregnancies. However small the risk is, the test cannot completely rule out the possibility of the baby having Down syndrome.

Why do you take my age into account?

Any woman may have a baby with Down syndrome, but the chance of this happening increases as a woman gets older. Therefore, we use age as one of the factors when assessing your risk of having a pregnancy with Down syndrome. Because older women are more likely to have a Down syndrome baby, they are also more likely to have a *screen positive* result.

For example, for women under the age of 35 about 4% will be screen positive, while for women who are 35 or older about 15% will be *screen positive*. Overall, about 5% of women will be *screen positive* and about 85% of Down syndrome pregnancies will be identified.

Can any other abnormalities be identified?

Yes, the risk of another chromosome abnormality, trisomy 18, can be estimated by the First Trimester Test.

Trisomy 18 (also known as Edwards syndrome) is a usually fatal abnormality caused by the presence of an extra chromosome number 18 in the cells of the developing baby. In the absence of screening, about 1 in every 7,000 babies is born with trisomy 18. The risk of trisomy 18 is reported only when the risk is 1 in 100 or higher. If your risk is high, you will be offered diagnostic testing. This is arranged by your health care provider. The first trimester screen detects about 6 out of 10 pregnancies affected with trisomy 18.

Another group of serious birth defects, called neural tube defects, are not tested for in first trimester screening. A little later on in your pregnancy, between 15 and 18 weeks, you may have a screening test for neural tube defects, which measures a substance called alpha-fetoprotein (AFP).

What are the tests that will be offered if my First Trimester Test result is screen positive?

If the test is *screen positive*, you will be offered a diagnostic test, usually chorionic villus sampling (CVS) or possibly an amniocentesis. The diagnostic test will determine whether or not the pregnancy is actually affected.

Chorionic Villus Sampling (CVS) - A CVS is offered early in pregnancy (usually between 10 and 13 weeks). CVS involves taking a sample of placental tissue (using local anaesthetic) either by inserting a needle through your abdominal wall or by passing a fine instrument through the cervix. CVS is performed under the guidance of an ultrasound scan and does not involve a stay in the hospital.

The CVS sample contains cells which can be used to tell whether or not the baby has Down syndrome or other chromosome problems. A result is usually ready within 1 to 2 weeks. There is a small risk associated with the CVS procedure. About 1% of women who have CVS will have a miscarriage as a result of the procedure.

Amniocentesis - Amniocentesis is a procedure in which the doctor obtains a small sample of fluid that surrounds the developing baby by passing a fine needle through the abdominal wall and into the uterus, under the guidance of an ultrasound scan. The fluid sample is then sent for laboratory testing to diagnose chromosome problems. A result is usually ready within 1 to 2 weeks. Amniocentesis is an invasive procedure, which means that there is a small risk of miscarriage (about 0.5% or 1 in 200) associated with it.

What happens if my baby does have Down syndrome?

A genetic counselor will be available to discuss your baby's diagnosis in detail and options available to you. One option would be to continue the pregnancy and make arrangements for appropriate medical services at and after delivery. Placing the infant for adoption after birth can also be considered. Another option would be termination of pregnancy.

Where can I get more information?

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