

# First Trimester Screening for Down Syndrome

## WHAT IS THE FIRST TRIMESTER SCREEN?

*The First Trimester Screen is a test used to calculate the risk for Down syndrome in a pregnancy. It can be performed as early as 11 weeks and as late as 13 weeks, 6 days counting from the first day of the last menstrual period. The result is usually available in one week and identifies 80% to 90% of cases of Down syndrome.*

*There are two parts to a First Trimester Screen: an ultrasound and a blood test. The ultrasound is used to measure the amount of fluid at the back of the fetal neck. This is called the nuchal translucency or NT. The NT is often larger in fetuses with Down syndrome. The blood test, which is performed from a simple finger stick, measures two proteins: Beta-hCG and PAPP-A. The amounts of these proteins are usually altered in Down syndrome pregnancies.*

*The chance for Down syndrome in the fetus is calculated by statistically combining the ultrasound, blood work results and patient's age. If the risk is as high as for a 35-year-old, the result falls into the positive range. About 5% of patients will have a positive result.*

### Are all pregnancies in the positive range affected?

No! In fact, most pregnancies with a “positive” result do not have a chromosome abnormality. Diagnostic tests (chorionic villus sampling or genetic amniocentesis) are offered to those with a “positive” result. With these tests, the fetus' chromosomes are studied from a small amount of tissue from the placenta or the fluid around the fetus.

### What is Down syndrome?

Down syndrome is caused by an extra chromosome. Symptoms include mental retardation and birth defects such as congenital heart disease. About one in every 800 babies is born with Down syndrome.

A fetus' risk for Down syndrome gets higher as the mother's age gets higher. This is why prenatal diagnosis is offered to women who are age 35 or older during pregnancy. However, most babies with Down syndrome are born to mothers in their twenties since women in this age group have the most births.

### Who should have the First Trimester Screen?

Anyone concerned about her risk to have a child with Down syndrome who is not already planning to have chorionic villus sampling or amniocentesis. This includes women

under age 35, who would not be offered invasive diagnostic testing, as well as those age 35 or older who do not wish to have invasive testing based upon their age-related risk alone.

Down syndrome screening is a way to predict which pregnancies are at higher risk so that prenatal diagnosis such as chorionic villus sampling and genetic amniocentesis can be offered appropriately.

### Are any other problems identified with the First Trimester Screen?

Yes. A high percentage of cases of Trisomy 18 will be identified with the first trimester screen. This common chromosome abnormality is usually fatal. Abnormal screen results may also indicate a risk for congenital heart disease or pregnancy complications.

### Does the First Trimester Screen detect spina bifida?

No. Screening for spina bifida must be done using alpha-fetoprotein (AFP) ONLY between 15 and 20 weeks of pregnancy, or with ultrasound at 18 to 22 weeks.

Women who have a first trimester screen SHOULD NOT have a triple or quad screen performed in the same pregnancy. This can result in a large number of false positive screens.

## Does insurance pay for first trimester screening?

Insurance companies have various policies regarding payment for the first trimester screen. Many consider the screen medically necessary for women who want information on the risk of having a child with Down syndrome. Perinatal center of Iowa bills for the ultrasound portion of the test, but sends all blood work for the first trimester screen to an outside laboratory, who will bill the insurance company directly. Please check with your own insurance company to find out if both parts of the test will be covered and at what rate. If you still have questions after speaking with your insurance provider or have other billing questions, you may reach our billing office staff at (515) 643-6888 or by email at [perinatalfinancial@mercydesmoines.org](mailto:perinatalfinancial@mercydesmoines.org).

## HOW CAN I SCHEDULE A FIRST TRIMESTER SCREEN?

Have your health care provider call Perinatal Center of Iowa at (515) 643-6888 to schedule an appointment.



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