

Answering Your Questions About: First Trimester Screening

What is first trimester screening?

First trimester screening (also called Ultra-Screen®) is a new screening option that helps determine a mother's risk for carrying a baby with Down syndrome (trisomy 21), trisomy 18 (Edwards syndrome) or trisomy 13 (Patau syndrome). The screening combines measurements from both a blood test and an ultrasound. The blood test involves measuring two substances in the mother's blood that are made by the pregnancy. These substances are called free beta-human chorionic gonadotrophin (β -hCG) and pregnancy-associated plasma protein A (PAPP-A). Both are produced as part of the baby's normal development. The ultrasound exam involves measuring the amount of fluid accumulation behind the baby's neck. This is known as the nuchal translucency (NT) measurement and has been shown to be helpful in identifying babies with chromosome conditions or other birth defects.

What if the first trimester screening results are abnormal?

It is important to remember that this is only a screening test, and not a diagnostic test. It is meant to identify pregnancies that may be at an increased risk for having a chromosomal abnormality. An abnormal screening does not mean that your baby has one of these conditions; it simply indicates that the risk is increased.

Follow-up is offered if results are abnormal. This follow-up may include genetic counseling, a detailed ultrasound, chorionic villus sampling (CVS), or amniocentesis. Your doctor will make the appropriate referral for follow-up.

What can first trimester screening identify?

Approximately 90 percent of babies with Down syndrome, trisomy 18 and trisomy 13 will have their condition detected through first trimester screening. In addition, babies with other types of chromosomal disorders or birth defects may also be identified through this screening. It is important to remember that a normal screening result does not eliminate the possibility that your baby could have one of these conditions, nor does it guarantee a normal baby.

What are Down syndrome, trisomy 18 and trisomy 13?

Down syndrome (trisomy 21), trisomy 18 and trisomy 13 are chromosome abnormalities. Chromosomes carry our genetic information. Typically, a person has 46 chromosomes in each cell of his or her body. Anytime a child receives extra or missing chromosomal material, developmental problems may occur.

An individual with Down syndrome has 47 chromosomes in each cell of his or her body because he or she has an extra copy of chromosome 21. Because of this extra chromosome, people with Down syndrome have some degree of mental retardation (typically mild to moderate). They also have a predisposition to certain medical conditions, such as heart defects.

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Babies with trisomy 18 and trisomy 13 also have 47 chromosomes in each cell of their bodies. These conditions are more severe than Down syndrome. Both conditions cause profound mental retardation and multiple birth defects. Few babies with either condition live more than one year, although some do survive into childhood. Like Down syndrome, the chance to have a baby with trisomy 18 or trisomy 13 increases with maternal age.

Women who are 35 or older at the time of delivery have an increased risk of having a baby with Down syndrome or other chromosome abnormality. This group of women is often referred for genetic counseling and ultrasound. They also have the option for diagnostic testing such as amniocentesis or CVS. Even though a woman is not considered to be at high risk until she reaches 35 years of age, babies with Down syndrome can be born to women of any age. The following table provides the age-related risk for having a child with Down syndrome and the risk to have a baby with any chromosome problem, including Down syndrome.

Maternal Age	Down syndrome risk	All chromosome abnormalities
25	1/1,205	1/476
30	1/885	1/384
35	1/365	1/178
40	1/109	1/63
45	1/32	1/18

How and when is this screening performed?

The screening involves drawing a sample of blood from the mother's arm to check levels of β -hCG and PAPP-A. The ultrasound examination measures the gestational age of the baby, along with the nuchal translucency (NT) measurement. The blood draw and ultrasound are generally done between 11 and 14 weeks of pregnancy. The result from the NT measurement is combined with the result from the blood test to generate a risk estimate for Down syndrome, trisomy 18 and trisomy 13.

Who should have first trimester screening?

Anyone who desires more information about her baby's development should consider screening. More than 95 percent of babies with Down syndrome, trisomy 18 and trisomy 13 are born to families with no history of these conditions. In some cases, the diagnosis of a problem before birth may lead to special management of the pregnancy and delivery to improve the outlook for the baby.

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