HARVEY J. STERN, MD, PhD, FACMG, FAAP

Dr. Stern joined GIVF in 1995 and is currently the Director of Reproductive Genetics and the GIVF Fetal Diagnostic Center. Dr. Stern has extensive experience in the evaluation of the fetus and newborn with congenital malformations and genetic syndromes. He is the primary physician involved in providing amniocentesis and chorionic villus sampling for GIVF's Prenatal Diagnosis program. He is board certified in Pediatrics and Medical Genetics with subspecialty certification in clinical, biochemical, and molecular genetics.

GENETIC COUNSELORS

Dr. Stern works closely with the genetic counselors in the Reproductive Genetics division at GIVF. Genetic counselors have a Masters degree in Genetics, a national board certification, and additional training in counseling and fetal development. They discuss information, provide support, and act as patient advocates throughout the genetic screening process. As the primary contacts for patients at GIVF, the genetic counselors help to coordinate testing and interpret results. In addition, genetic counselors specialize in reviewing family histories and assessing risk factors that may affect pregnancies or other family members.

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GENETICS & IVF INSTITUTE

First Trimester Screening

The first trimester screen (FTS) is a non-invasive way to estimate the chance for three common chromosome abnormalities in pregnancy.

Trisomy 21, also known as Down syndrome, is associated with mild to moderate intellectual disability, heart problems, muscle weakness, and other birth defects.

Trisomy 18 and **Trisomy 13** are associated with severe intellectual disability and birth defects. These pregnancies usually result in miscarriage, stillbirth, or infant loss.

Any pregnancy could have one of these three conditions, even when the family history is normal.

WHO SHOULD CONSIDER THE FTS?

The FTS is recommended for pregnant women of any age who would like early information about the health of a pregnancy without invasive testing. Serious chromosomal abnormalities are more commonly associated with pregnancies in women age 35 and older, but they can occur at any age. In fact, more babies with Down syndrome are born to women under age 35.

WHAT IS THE FTS?

The FTS is a non-invasive tool that combines a blood test, a specialized ultrasound, and your age. This information provides a powerful, personalized risk assessment for your current pregnancy. It detects up to:

- 98% of pregnancies with Down syndrome, and
- 95% of pregnancies with Trisomy 18 or Trisomy 13.

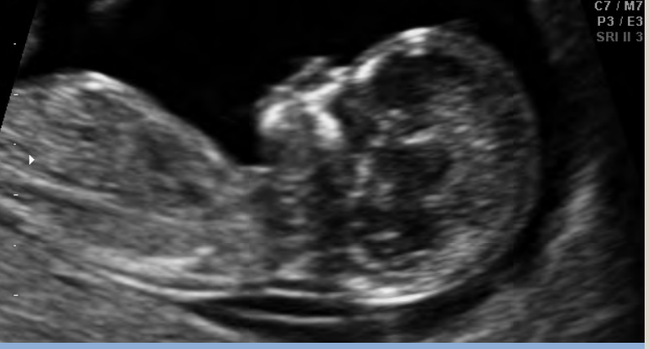
This screening process is highly accurate and available early in pregnancy. However, no screen can assess the risk for every chromosome abnormality, genetic disease, or birth defect.

THE FTS OFFERS EVERY PREGNANT WOMAN:

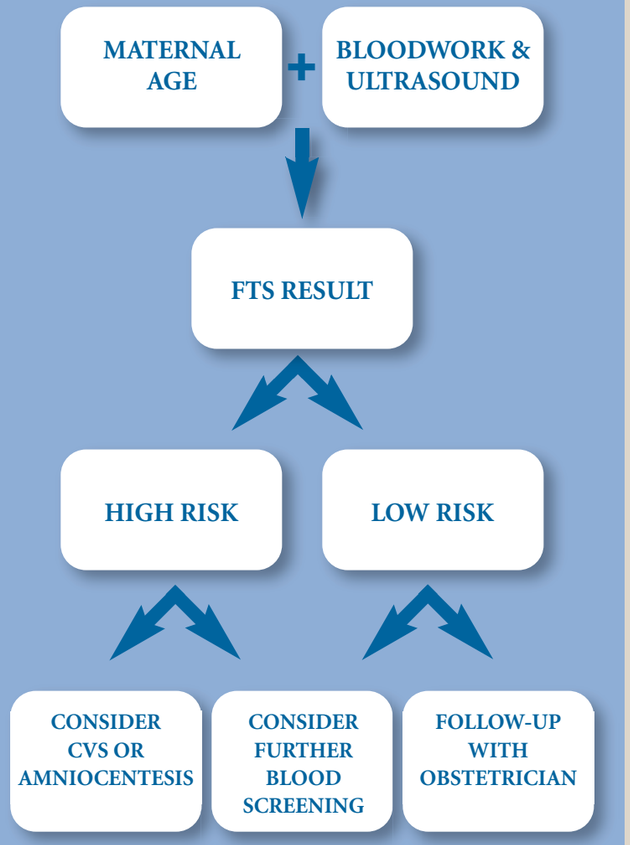
- A personalized risk assessment instead of a generic risk based only on your age.
- A highly accurate non-invasive screening available in pregnancy.
- Earlier information than second trimester options.
- An individualized risk for each twin or triplet in a multiple gestation pregnancy.

IF YOU ARE 35 AND OVER, THE FTS ALSO OFFERS YOU:

- A non-invasive first step to determine if diagnostic testing is appropriate for you.
- Additional information to help you choose between follow-up options.



FIRST TRIMESTER SCREENING STEPS



HOW IS THE FTS PERFORMED?

At your FTS appointment, you will have an ultrasound and a blood draw. The blood sample is used to measure the concentration of five proteins, PAPP-A (pregnancy associated plasma protein A), free β-hCG (free-beta human chorionic gonadotropin), AFP (alpha-feto-protein, DIA (Dimeric Inhibin A), and PGIF (Placental Growth Factor). These proteins are released into the maternal blood in every pregnancy. The ultrasound looks for the presence of the fetal nasal bone and measures the nuchal translucency. The nuchal translucency is a fluid filled area at the back of the fetal neck, present in every pregnancy.

Information from the blood test and ultrasound are combined with your age to generate your result. The personalized results will describe the risks for Down syndrome and for Trisomy 18 or Trisomy 13 in your pregnancy.

WHEN SHOULD I HAVE MY FTS?

The FTS ultrasound is performed during a specific window of time based on fetal size. For most women, the FTS occurs in the 12th or 13th week of pregnancy. The bloodwork can be done at the same time, or in the two weeks prior to the ultrasound. For convenience, most women will have their ultrasound and bloodwork completed at the same appointment.

HOW WILL I GET MY FTS RESULTS?

The Genetics & IVF Institute (GIVF) has on-site genetic counselors who will meet with you the day of your ultrasound and call you with your FTS results, generally less than a week after your appointment. Genetic counselors have extensive training in prenatal screening and a thorough understanding of the test implications; they will explain the results to you and answer your questions.

WHAT WILL MY FTS RESULTS TELL ME?

The genetic counselor will give you two results. The first result is the chance for Down syndrome. The second result is the chance for Trisomy 18 or Trisomy 13. These results are expressed as probabilities, such as 1 in 1000 (low risk) or 1 in 100 (high risk). The genetic counselor will explain your personalized results and what they mean for your pregnancy.

Most women's results indicate low risk and provide reassurance that further screening or invasive testing for chromosome abnormalities is probably not necessary. Some women may be offered further blood screening options. Other women will be offered diagnostic follow-up testing, such as chorionic villus sampling (CVS) or amniocentesis.

All follow-up options are elective. You will be able to decide what next steps (if any) are right for you based on your FTS results, pregnancy history, and personal preferences.

WHAT ELSE COULD MY FTS TELL ME?

Enlargement of the nuchal translucency indicates that the fetus has an increased risk for chromosome abnormalities. An enlarged nuchal translucency can also be seen in some pregnancies with a heart defect or a rare genetic disorder. Additional testing or follow-up procedures may be recommended for later in pregnancy.

WHY SHOULD I HAVE MY FTS AT GIVF?

- For your convenience, GIVF gives you the option of completing the bloodwork and ultrasound on the same day. Alternative scheduling can be arranged.
- You will meet with a genetic counselor on the day of your appointment. The counselor will answer your questions about the screening and inform you of your ultrasound findings. She will also speak to you about your genetic family history.
- The genetic counselor will call you directly with your FTS results. This is another opportunity to ask questions and review possible follow-up options based on the ultrasound or family history information.
- Patients with abnormal results have immediate access to genetic counselors and often same day diagnostic testing if necessary.
- GIVF provides the complete range of patient support, non-invasive screening, and diagnostic testing. This continuity of care ensures you get complete and accurate information as quickly as possible.
- The FTS is only accurate when performed by certified professionals. The staff of GIVF is highly skilled and certified by The Fetal Medicine Foundation.

HOW TO SCHEDULE AN APPOINTMENT

Contact GIVF early in your pregnancy to schedule an appointment.

(703) 698-7355
www.givf.com
givf@givf.com

You will be asked to provide the due date given to you by your obstetrician when scheduling your appointment.