Northwestern Reproductive Genetics, Inc.
Eugene Pergament, MD, PhD, FACMG

GENERAL INFORMATION ABOUT FIRST TRIMESTER SCREENING

What is First Trimester Screening?

First Trimester Screening is a first-trimester prenatal screening procedure that provides specific risk assessments for Down syndrome (trisomy 21) and trisomies18 and13. The First Trimester Screen combines ultrasound measurement of the amount of fluid behind the neck of a fetus (nuchal translucency) with maternal serum markers (free beta hCG and PAPP-A). It is the earliest and most reliable Down syndrome screen available, with a detection rate of 91% and an overall false positive rate of 5%. The First Trimester Screen also detects 95% of trisomies 18 and 13 pregnancies with a 0.3% overall false positive rate.

Who should be screened?

Leading medical professional organizations recommend first trimester Down syndrome screening for all pregnant women, regardless of age.

Women age 35 and older and those with a family history of Down syndrome are more likely to have a pregnancy affected by Down syndrome. However, most cases of Down syndrome occur in women younger than 35.

At what gestational age should the First Trimester Screen be performed?

First Trimester Screening consists of two parts: a blood test that can be done between 9-13 weeks, and an ultrasound examination performed between 11-14 weeks in pregnancy. We generally schedule our patients for their blood draw during their 11th week in pregnancy and for their ultrasound during their 12th week.

What are the advantages of doing First Trimester Screening?

- For those shown to be at low risk, First Trimester Screening reduces the use of unnecessary invasive diagnostic procedures that increase health risks to patient and fetus
- For those shown to be at high risk, First Trimester Screening gives the patient and physician more time to consider follow-up diagnostic options such as CVS (chorionic villus sampling) or amniocentesis
- First Trimester Screening is safe, simple, and non-invasive

CONDITIONS INCLUDED IN FIRST TRIMESTER SCREENING

Down syndrome

Down syndrome, also known as trisomy 21, is a congenital disorder caused by the presence of an extra copy of chromosome 21, in which the affected person has mild to moderate mental retardation, characteristic physical appearance, and often has congenital malformations such as heart defects.

Trisomy 18

Trisomy 18 is a congenital disorder caused by the presence of an extra copy of chromosome 18, occurring in about 1 out of 3,000 to 1 out of 6,000 live births. Trisomy 18 is generally fatal, with 50% of babies dying within the first week of life and an additional 40% dying within the first year of life. The disorder is characterized by profound mental retardation, heart defects, and central nervous system defects.

Trisomy 13

Trisomy 13 is a **congenital disorder** caused by the presence of an extra copy of chromosome 13, occurring in about 1 out of 10,000 live births. Trisomy 13 is generally fatal, with more than 80% of babies dying in the first month of life. The disorder is characterized by profound mental retardation, cardiac problems, and congenital abnormalities.

It is important to note that First Trimester Screening cannot detect all birth defects and genetic syndromes.

WHAT TO EXPECT DURING YOUR GENETICS CONSULTATION

During your genetics consultation, we will review all aspects of First Trimester Screening, including details of the test and options for further testing, if indicated. We will also review your options for ethnicity-based genetic carrier screening during your visit with us.

If you were able to have your blood drawn the week prior to your ultrasound, we will be able to review the results of your First Trimester Screen during your genetics consultation.

UNDERSTANDING YOUR RESULTS

What does a "screen negative" result mean?

A screen negative result means that the risks for Down syndrome, trisomy 13, and trisomy 18 are low enough that diagnostic testing (CVS or amniocentesis) is *not* indicated. The majority of pregnant women have a "screen negative" result. Although a "screen negative" result greatly reduces the risk for the screened chromosome abnormalities, it does not completely rule out the possibility that the fetus is affected.

If your First Trimester Screen is "screen negative", your obstetrician will provide you with routine obstetrical care. This will likely include undergoing second trimester maternal serum screening, which is generally performed between 15-18 weeks in pregnancy. After results are obtained from your second trimester maternal serum screen, a combined screen result (United Screening Approach) can be calculated, sent to you in the mail, and faxed to your obstetrician. The USA provides risk assessments for a number of chromosome abnormalities, as well as an *overall* risk for *any* chromosome abnormality in the pregnancy.

What does a "screen positive" result mean?

A screen positive result means that there is an increased risk for Down syndrome, trisomy 13, or trisomy 18 in the pregnancy. A screen positive result does *not* mean that the fetus is affected with the condition, only that a pregnancy has an increased risk for the condition. Most women with "screen positive" results have healthy babies.

When the result is "screen positive", the following options are available:

- Diagnostic testing by chorionic villus sampling (CVS) performed between 10-13 weeks
- Diagnostic testing by amniocentesis performed after 15 weeks
- Second trimester maternal serum screening drawn after 15 weeks to obtain a combined first and second trimester screening result (USA)
- Level II ultrasound performed at approximately 20 weeks

QUESTIONS?

If you have any questions concerning the above information, please do not hesitate to contact us at 312-981-4400.