

INTRODUCTION

Most babies are born normal. All women, however, have a very small chance of having a baby with a chromosome problem such as Down Syndrome. This risk is related to the mother's age at delivery. Traditionally Down Syndrome screening has been performed at 16-20 weeks of pregnancy. Recently, a test performed between 11 and 13 weeks has been introduced that provides women with reassurance early in their pregnancy that they are at low risk for this problem.

WHAT IS FIRST TRIMESTER SCREENING?

It is a combination of a finger stick blood test (the serum screen) and a specialized ultrasound exam performed in the early part of pregnancy to assess a woman's individual risk for Down Syndrome and Trisomy 18.

THE SERUM SCREEN

This blood test can be taken between 11 and 13 weeks of pregnancy. Two chemicals normally found in the blood during pregnancy are analyzed by the laboratory: Beta hCG and PAPP-A.

THE FIRST TRIMESTER ULTRASOUND

This ultrasound is performed between 11 and 13 weeks. Measurements will be taken to confirm the gestational age of the baby and you can see the heart beat. It is difficult to see all structures this early, but a general survey will be performed to reassure you the baby appears normal. The nuchal translucency(NT) or amount of fluid behind the fetal neck is measured. This NT measurement is combined with the blood test result to assess your risk for Down Syndrome and Trisomy 18.

HOW ACCURATE IS FIRST TRIMESTER SCREENING?

This combination of the blood test and NT will detect 90% of fetuses with Down Syndrome. Many fetuses with Trisomy 18 will also be found by this method. In some cases, the First Trimester Screen detects other abnormalities as well. The test cannot exclude certain birth defects and developmental delays.

WHO SHOULD HAVE FIRST TRIMESTER SCREENING?

Any pregnant woman seeking early reassurance about her baby's health may consider First Trimester Screening.

WHAT WILL HAPPEN DURING MY VISIT?

You will meet with a genetic counselor to briefly discuss first trimester screening. She will then obtain a blood sample from your finger for the serum screen. A sonographer and physician will perform your ultrasound. **You do not need a full bladder for this exam.** You will be given results of the ultrasound during your visit. The genetic counselor will call you within a week to discuss your First Trimester Screen results. If you have additional concerns after the screening is completed, you may schedule a follow up consultation to discuss these issues in more detail.

WHAT IF I HAVE ADDITIONAL CONCERNS?

Some women have a personal or family history of a genetic problem for which they would like more information and possibly testing. A genetic counselor can arrange a consultation before your First Trimester Screening appointment to discuss these issues in detail.

WHAT IF MY RESULTS SHOW A LOW RISK?

Most women will be given reassuring test results. For those younger than 35 years at delivery, a maternal serum alpha fetoprotein(MSAFP-a blood test) at 15-20 weeks and a screening ultrasound at 18-20 weeks are recommended. Women who will be 35 or older should also have the MSAFP as well as a targeted ultrasound examination. The option of an integrated screen(blood test in first and second trimester), recently introduced, is also available to all women.

WHAT IF MY RESULTS SHOW INCREASED RISK?

An Increased risk does not mean your baby has a problem. It means you should consider additional evaluation and/or testing. If your results show an increased risk, the genetic counselor will explain your results and your options may include chorionic villus sampling or amniocentesis to examine the baby's chromosomes directly but this decision will be individualized following the counseling.



SUMMARY OF PRENATAL DIAGNOSIS OPTIONS

Screening Tests

- First Trimester Screen(11-13 weeks) followed by MSAFP(16-18 weeks)
- Second Trimester Quad Screen (16-20 weeks)
- Integrated Screening (First and Second Trimester testing)
- Screening Ultrasound for women younger than 35 at delivery (18-20 weeks)
- Targeted Ultrasound for women 35 and older at delivery (18-20 weeks)

Genetic Testing

- Chorionic Villus Sampling (11-13 weeks)
- Amniocentesis (15-20 weeks)

FIRST TRIMESTER SCREENING PROGRAM



Saint Alphonus

Maternal-Fetal Medicine

900 N. Liberty, Suite 206
Boise, ID 83706
(208) 367-5544



Saint Alphonus

Maternal-Fetal Medicine