

First Trimester Screening **Patient Information**

What is first trimester screening?

First trimester screening is a prenatal screening test for Down syndrome, trisomy 18 and trisomy 13. Down syndrome, trisomy 18 and trisomy 13 are chromosome abnormalities that can cause significant health and developmental problems for your baby. Chromosomes are the packages of genetic information in our bodies' cells. Down syndrome is the most common chromosome abnormality. It occurs in about 1 out of every 800 newborn babies. Although it is possible for a woman of any age to have a baby with a chromosome abnormality, the chance is greater as she gets older. Most chromosome abnormalities are not hereditary (they do not usually run in families).

Can first trimester screening tell me if my baby has a chromosome abnormality?

No. First trimester screening is a **screening** test. This means the test can only tell you how **likely** it is that the baby **could** have one of these conditions. First trimester screening **cannot** tell you if the baby actually has a chromosome abnormality or not. First trimester screening will not detect about 1 out of every 5 pregnancies with Down syndrome.

When and how is the screening test done?

First trimester screening is usually done between 9 and 13 weeks of pregnancy. It involves a blood test and an early ultrasound measurement of the back of the neck of the developing baby (the "nuchal translucency" or NT). The results of these tests are combined with your age to estimate your chance of having a baby with Down syndrome, and trisomy 18 or trisomy 13.

Results are given as a numerical risk estimate, such as a "1 in 200 risk for Down syndrome" or a "1 in 10 risk for trisomy 18". The test result is considered "screen positive" if the risk estimate is higher than a certain cutoff point. For Down syndrome, a risk of greater than about "1 in 300" is "screen positive".

What if I have a "screen positive" test result?

Women with screen positive test results are referred for genetic counseling to discuss available options. Chorionic villus sampling (CVS) and amniocentesis are two diagnostic tests that can determine if the baby has a chromosome abnormality or not. These tests have a small risk of miscarriage.

What are the advantages of first trimester screening?

- ✓ Screen positive first trimester screening results may be available in time for CVS (an earlier diagnostic test than amniocentesis).
- ✓ Some studies suggest that the detection rate for Down syndrome using first trimester screening may be slightly higher than the standard second trimester screening test, the California Expanded AFP test.

What are the disadvantages of first trimester screening?

- ✓ First trimester screening is only available at select non-Kaiser Permanente facilities.
- ✓ It does not screen for other conditions such as neural tube defects, abdominal wall defects, and Smith Lemli-Opitz syndrome (all included in second trimester Expanded AFP screening).
- ✓ First trimester screening cannot be completed in some women. These women may choose to have Expanded AFP screening.
- ✓ First trimester screening results may not be available in time for CVS. These women with screen positive results may have a longer period of anxiety while waiting to have amniocentesis.
- ✓ CVS is a procedure that:
 - has a higher risk of miscarriage (about 1 in 100) than amniocentesis (about 1 in 400).
 - cannot be done in all patients
 - has higher rate of unclear results than amniocentesis

Women over 35 years of age

Some women 35 years of age and older at the time of delivery may not want to have any screening tests. They may choose to proceed directly to a diagnostic test such as CVS or amniocentesis. A genetic counselor can meet with you to discuss your options.

A Comparison of First Trimester Screening and Second Trimester Screening

You may choose to have one test or the other. You may also choose to have **no** screening.

Having both tests is **not** recommended because, at this time, these tests are designed to be offered alone. Having both tests as currently designed may result in too many women having diagnostic tests, which would result in more miscarriages.

	First Trimester Screening (FTS)	Second Trimester Screening (Expanded AFP testing)
Screens for	<ul style="list-style-type: none"> • Down syndrome • trisomy 18 & trisomy 13 	<ul style="list-style-type: none"> • Down syndrome • trisomy 18 • neural tube defects • abdominal wall defects • Smith-Lemli-Opitz syndrome
Test will	<ul style="list-style-type: none"> • Identify about 1 out of 20 pregnancies as being “screen positive” (at increased risk) for Down syndrome • Detect about 4 out of 5 pregnancies with Down syndrome • Not detect about 1 out of 5 pregnancies with Down syndrome 	<ul style="list-style-type: none"> • Identify about 1 out of 20 pregnancies as being “screen positive” (at increased risk) for Down syndrome • Detect about 4 out of 5 pregnancies with Down syndrome • Not detect about 1 out of 5 pregnancies with Down syndrome
Type of test	<ul style="list-style-type: none"> • Blood test & early ultrasound 	<ul style="list-style-type: none"> • Blood test
Timing	<ul style="list-style-type: none"> • 9-13 weeks of pregnancy for blood test • 11-13 weeks of pregnancy for NT ultrasound 	<ul style="list-style-type: none"> • 15-20 weeks of pregnancy
Follow-up options for diagnostic testing if result is “Screen Positive”	<ul style="list-style-type: none"> • CVS, if the results are available early (miscarriage risk about 1 in 100). (CVS cannot be done in all patients). • Amniocentesis if the results are available later 	<ul style="list-style-type: none"> • Detailed ultrasound and amniocentesis (miscarriage risk about 1 in 400).
Wait time between “Screen Positive” result and a diagnostic follow-up test (CVS or amniocentesis)	<ul style="list-style-type: none"> • A few days to a few weeks 	<ul style="list-style-type: none"> • 7 days or less
Additional testing later in pregnancy	<ul style="list-style-type: none"> • Routine ultrasound • MSAFP-only (maternal serum AFP) blood test to screen for neural tube defects 	<ul style="list-style-type: none"> • Routine ultrasound

Deciding which screening test, if any, you want to have is a personal decision. Talk with your doctor, nurse practitioner, nurse midwife or genetic counselor for more information.

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