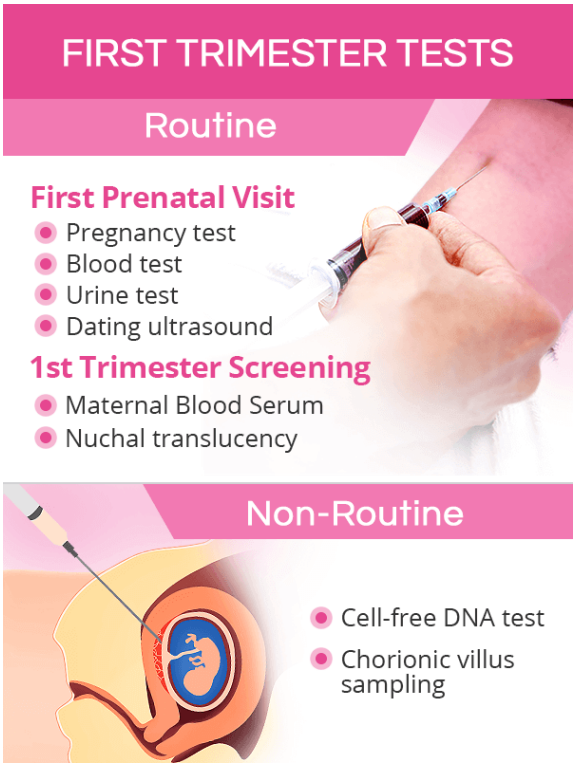
Prenatal Screening

Prenatal Testing



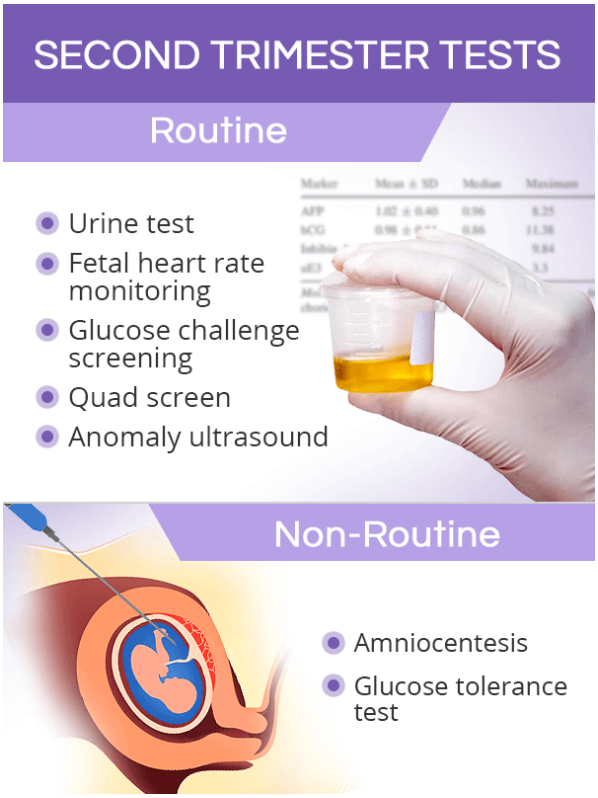
First trimester screening includes:

- **Ultrasound for fetal nuchal translucency**. Nuchal translucency screening uses an ultrasound to examine the area at the back of the fetal neck for increased fluid or thickening.
- **Ultrasound for fetal nasal bone determination**. The nasal bone may not be visualized in some fetuses with certain chromosome abnormalities, such as Down syndrome. This screen is performed using an ultrasound between 11 and 13 weeks gestation.
- **Maternal serum (blood) tests**. These blood tests measure two substances found in the blood of all pregnant women:
    - **Pregnancy-associated plasma protein A**. A protein produced by the placenta in early pregnancy. Abnormal levels are associated with an increased risk of chromosomal abnormality.
    - **Human chorionic gonadotropin**. A hormone produced by the placenta in early pregnancy. Abnormal levels are associated with an increased risk of chromosomal abnormality.

When used together as first trimester screening tests, nuchal translucency screening and maternal blood tests have a greater ability to determine if the fetus might have a birth defect, such as Down syndrome (trisomy 21) and trisomy 18.

Prenatal Testing

If the results of these first trimester screening tests are abnormal, genetic counseling is recommended. Additional testing, such as chorionic villus sampling, amniocentesis, cell-free fetal DNA, or other ultrasounds, may be needed for an accurate diagnosis.



Second Trimester Prenatal Screening Tests

Second trimester prenatal screening may include several blood tests called multiple markers. These markers provide information about the potential risk of having a fetus with certain genetic conditions or birth defects. Screening is usually done by taking a sample of the blood between 15 and 20 weeks of pregnancy (16 to 18 weeks is ideal). The multiple markers include:

- **AFP screening**. Also called maternal serum AFP, this blood test measures the level of AFP in the blood during pregnancy. AFP is a protein normally produced by the fetal liver that is present in the fluid surrounding the fetus (amniotic fluid). It crosses the placenta and enters the blood. Abnormal levels of AFP may indicate:
    - A miscalculated due date, as the levels vary throughout pregnancy
    - Defects in the abdominal wall of the fetus
    - Down syndrome or other chromosomal abnormalities
    - Open neural tube defects, such as spina bifida
    - Twins (more than one fetus is producing the protein)
- **Estriol**. This is a hormone produced by the placenta. It can be measured in maternal blood or urine to be used to determine fetal health. Low estriol (uE3) levels may be the

Prenatal Testing

result of fetal demise, congenital abnormalities, or some genetic hormonal disorders of the fetus.

- **Inhibin**. This is a hormone produced by the placenta. Elevated Inhibin A levels in pregnancy are significantly associated with pre-eclampsia, GDM, macrosomia, low birth weight and preterm delivery.
- **Human chorionic gonadotropin**. This is also a hormone produced by the placenta. Quantitative HCG measurement helps determine the exact age of the fetus.

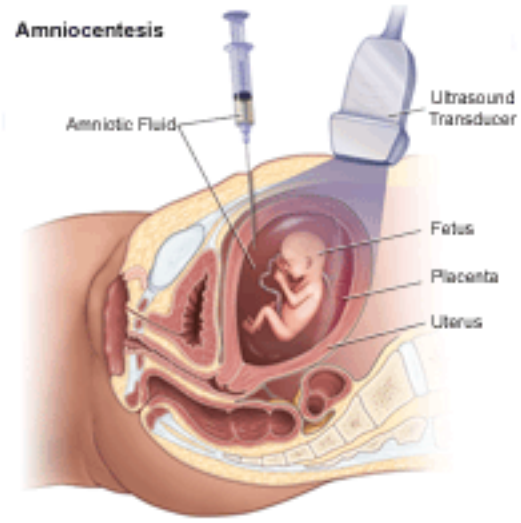
Abnormal test results of AFP and other markers may mean that additional testing is needed. An ultrasound is used to confirm the milestones of the pregnancy and to check the fetal spine and other body parts for defects. An amniocentesis may be needed for an accurate diagnosis.

Since multiple marker screening is not diagnostic, it is not 100 % accurate. It helps determine who in the population should be offered additional testing during pregnancy.

When pregnant patients have both first and second trimester screening tests performed, the ability of the tests to detect an abnormality is greater than using just one screening independently. Most cases of Down syndrome can be detected when both first and second trimester screenings are used.

Amniocentesis

An amniocentesis involves taking a small sample of the amniotic fluid that surrounds the fetus. It is used to diagnose chromosomal disorders and open neural tube defects, such as spina bifida. Testing is available for other genetic defects and disorders depending on the family history and the availability of lab testing at the time of the procedure.



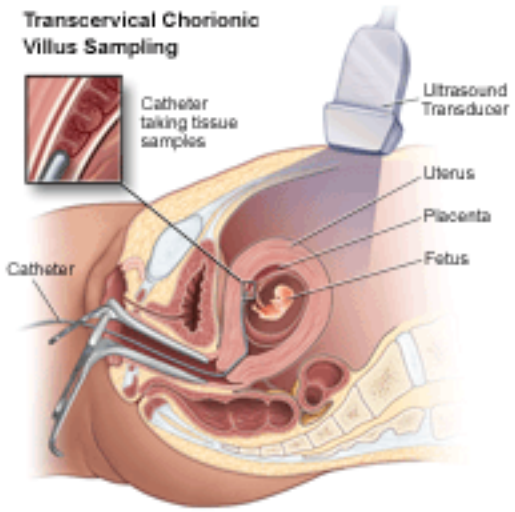
An amniocentesis is generally offered to women between the 15th and 20th week of pregnancy who have an increased risk of chromosomal abnormalities. Candidates include women who will be over age 35 at the time of delivery or those who have had an abnormal maternal serum screening test.

Prenatal Testing

An amniocentesis involves inserting a long, thin needle through the abdomen into the amniotic sac to withdraw a small sample of amniotic fluid. The amniotic fluid contains cells shed by the fetus, which contain genetic information.

Women who are pregnant with twins or other higher-order multiples need sampling from each amniotic sac to study each fetus. Depending on the position of the fetus and placenta, amount of fluid, and woman's anatomy, sometimes the amniocentesis cannot be done. The fluid is then sent to a genetics lab so that the cells can grow and be analyzed. AFP is also measured to rule out an open neural tube defect. Results are usually available in about 10 days to two weeks, depending on the lab.

Chorionic Villus Sampling (CVS)



CVS is a prenatal test that involves taking a sample of some of the placental tissue. This tissue contains the same genetic material as the fetus and can be tested for chromosomal abnormalities and some other genetic problems. Testing is available for other genetic defects and disorders, depending on the family history and the availability of lab testing at the time of the procedure. Unlike amniocentesis, CVS does not provide information on open neural tube defects. Therefore, women who undergo CVS also need a follow-up blood test between 16 and 18 weeks of pregnancy to screen for these defects.

CVS may be offered to women with an increased risk of chromosomal abnormalities or who have a family history of a genetic defect that is testable from the placental tissue. CVS is usually performed between the 10th and 13th week of pregnancy. Although exact methods may vary, the procedure involves the following steps:

- The physician will insert a small tube (catheter) through the vagina and into the cervix.
- Using ultrasound technology, the physician will guide the catheter into place near the placenta and will remove some tissue using a syringe on the other end of the catheter.

Prenatal Testing

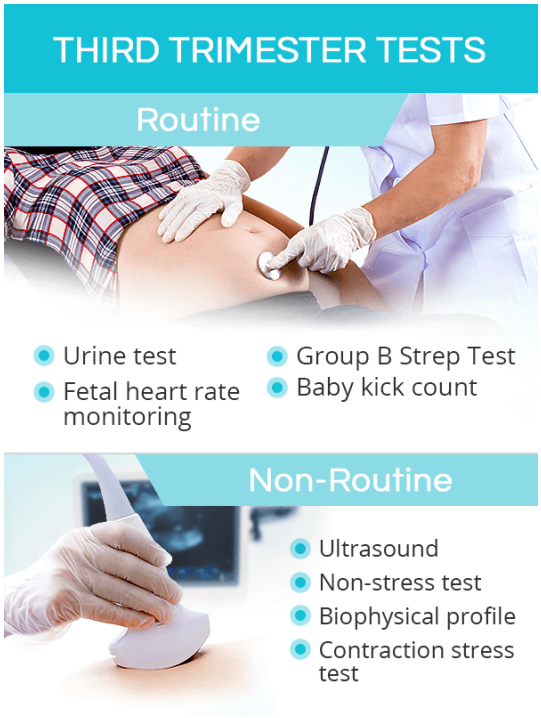
The physician may also choose to perform a transabdominal CVS, which involves inserting a needle through the abdomen and into the uterus to sample the placental cells. The tissue samples are sent to a genetic lab for growth and analysis. Results are usually available in about 10 days to two weeks, depending on the lab.

Women with twins or other higher-order multiples usually need sampling from each placenta. However, because of the complexity of the procedure and the positioning of the placentas, CVS is not always feasible or successful with multiples.

Women who are not candidates for CVS or who did not get accurate results from the procedure may require a follow-up amniocentesis. An active vaginal infection, such as herpes or gonorrhea, will prohibit the procedure. In other cases, the physician may take a sample that does not have enough tissue to grow in the lab, generating incomplete or inconclusive results.

Glucose Testing

A glucose challenge test is usually conducted between 24 and 28 weeks of pregnancy. Abnormal glucose levels may indicate gestational diabetes. The initial one-hour test is a glucose challenge test. If the results are abnormal, a glucose tolerance test is needed. The patient may be asked to only drink water on the day the glucose tolerance test is given.



Group B Strep Culture

Group B streptococcus (GBS) is a type of bacteria found in the lower genital tract of about 20 % of all women. While a GBS infection does not usually cause problems in women before pregnancy, it can cause serious illness in patients during pregnancy. GBS may cause

Prenatal Testing

chorioamnionitis (a severe infection of the placental tissues) and postpartum infection. Urinary tract infections caused by GBS can lead to preterm labor and birth or pyelonephritis and sepsis.

GBS is the most common cause of life-threatening infections in newborns. Newborns contract the infection during pregnancy or from the mother's genital tract during labor and delivery.

The Centers for Disease Control and Prevention recommends screening all pregnant women for vaginal and rectal GBS colonization between 35 and 37 weeks gestation. The treatment of pregnant patients with certain risk factors or positive cultures is important to reduce the risk of transmission of GBS to the fetus. Infants whose mother receive antibiotic treatment for a positive GBS test are 20 times less likely to develop the disease than those without treatment.