

Although most babies are born healthy, the risk of producing a child with a birth defect concerns many parents. Risks for certain types of birth defects, such as open neural tube defects and Down syndrome, can be assessed by screening tests. Approximately 5 percent of those undergoing screening are determined to be at increased risk and further testing can be performed to achieve a definitive diagnosis.

In 1985, maternal serum alpha-fetoprotein (AFP) screening for open neural tube defects became a standard of second trimester obstetric care. Since this time, additional chemical markers have been added to this testing in the second trimester, enabling screening for chromosome abnormalities like Down syndrome and trisomy 18, a condition resulting in severe mental retardation, physical birth defects, and usually death shortly after delivery.

In 2004, first trimester screening for Down syndrome and trisomy 18 emerged, providing patients with a variety of screening options. The information provided by screening tests is beneficial for some families; other families do not feel this information is helpful to have prior to delivery. This brochure is intended to help you explore these options. Participation in the screening tests is your choice. Your obstetrician or a genetic counselor can also provide you with more information.

If you have questions about screening or diagnostic tests in pregnancy, please call the MUSC Prenatal Wellness Center at 792-1212.

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If you are a patient, please contact your obstetrician or call the **MUSC Prenatal Wellness Center** at

1-800-424-MUSC

or **792-1212.**

If you are a physician, please call through MEDLINE at

1-800-922-5250 or 792-2200.

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**Prenatal
Wellness Center**

**Prenatal Screening
and Diagnosis**

A guide for expectant women

First trimester screening

(Combined screening)

First trimester screening is performed between 11-13 weeks of pregnancy, and involves both an ultrasound and a blood test.

Ultrasound

Ultrasound measures an area of fluid accumulation at the back of the baby's neck called nuchal translucency (NT). This accumulation of fluid is a normal finding. Increased NT measurements, however, may indicate increased risk for chromosome abnormalities, congenital heart defects, and other genetic syndromes.

Blood test

The blood test by a fingerstick measures the levels of two proteins, freeBeta-hCG and PAPP-A, which are normally found in the blood of pregnant women. The levels of these two chemicals are combined with the NT measurement and the mother's age to provide a risk assessment for Down syndrome and trisomy 18.

The combined testing detects approximately 85 percent of Down syndrome and 97 percent of trisomy 18.

Patients determined to be at increased risk for either of these conditions can then be offered diagnostic testing such as chorionic villus sampling (CVS) or amniocentesis. If the screening test does not predict an increased risk, it is recommended that the patient proceed with maternal serum AFP screening in the second trimester to identify increased risks for open neural tube defects and abdominal wall defects.

It is important to remember that a normal screening test does not guarantee a normal baby, nor do abnormal test results definitely mean a baby with a birth defect.

Second trimester screening

Second trimester screening is performed between 15-21 weeks of pregnancy. A blood test measures the levels of four substances normally found in the blood of pregnant women: AFP, hCG, estriol, and inhibin A. Based on the levels of these four substances and the mother's age, a risk assessment is provided for open neural tube defects, Down syndrome and trisomy 18.

The screening can detect approximately 80 percent of open neural tube defects, 80-85 percent of Down syndrome, and 60 percent of trisomy 18.

Women with an increased risk for any of these conditions from this screening will be offered further testing.

Women older than 35 at delivery

Any woman at any age can have a baby with a birth defect, but the risk for chromosome abnormalities increases with age. The American College of Obstetricians and Gynecologists and the American College of Medical Genetics recommend that all women who are 35 years of age and above at delivery be offered chorionic villus sampling (CVS) or amniocentesis for prenatal chromosome testing to diagnose 99.9 percent of chromosome abnormalities.

Chorionic villus sampling

CVS is performed between 10-12 weeks of pregnancy and involves the removal of placental tissue for chromosome analysis.

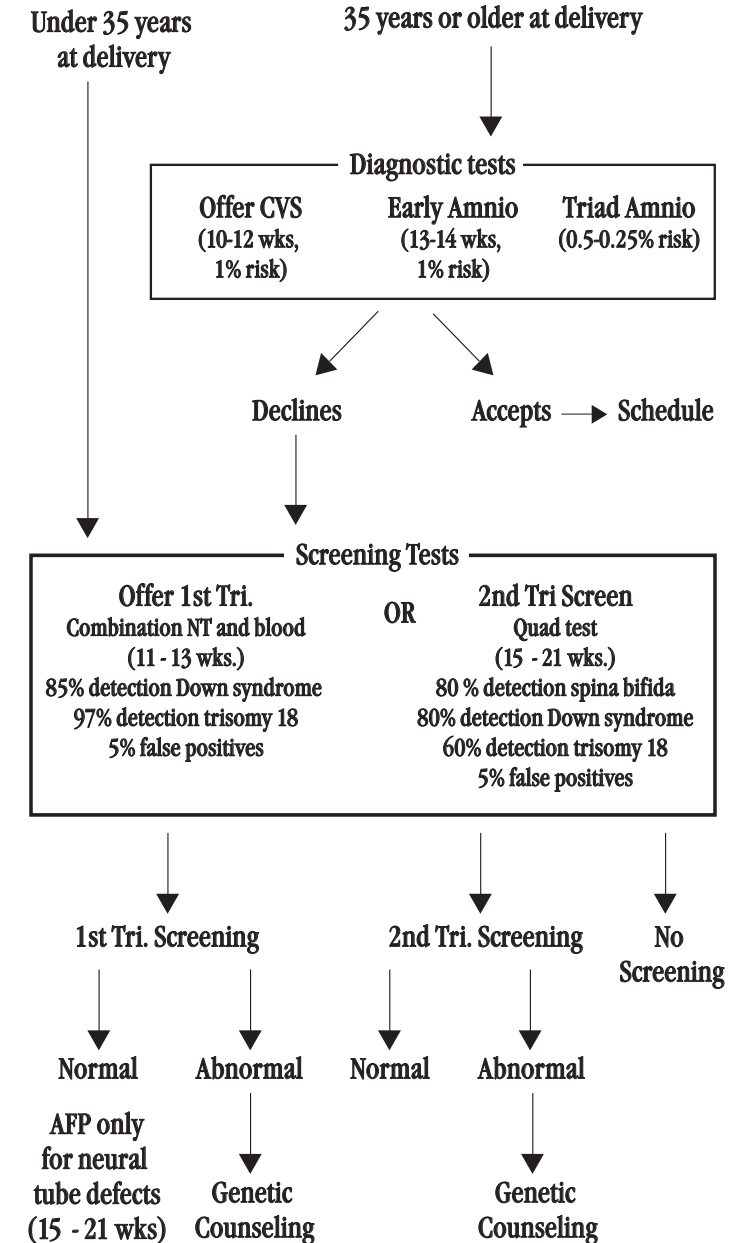
Amniocentesis

Amniocentesis involves removal of a small amount of amniotic fluid. Chromosome analysis is performed on the baby's skin cells in the amniotic fluid. Amniocentesis can be performed between 13-14 weeks in pregnancy (early amniocentesis) or at 15 weeks and later (traditional amniocentesis).

Both CVS and amniocentesis carry an increased risk for pregnancy complications including miscarriage. The risk with CVS is approximately 1 percent. The risk with early amniocentesis is also approximately 1 percent. The risk with traditional amniocentesis is 0.5 percent or less.

Some women over 35 will choose to decline diagnostic testing for a variety of reasons and opt to proceed with a screening test. While this can be done, it is important to remember that screening tests are not a substitute for diagnostic tests. Screening tests do not provide the full information that diagnostic testing does, because they screen for only two of the chromosome abnormalities for which there is an increased risk. Additionally, normal screening results may falsely reassure women who still have an increased risk for chromosome abnormality due to age.

Prenatal Screening & Diagnosis



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