

## (special prenatal tests

Even though your screening results may be atypical, it does not mean your baby has a birth defect.

If the doctor has questions or concerns based on your screening results and medical history, he or she will likely offer prenatal diagnostic tests such as amniocentesis or chorionic villus sampling (CVS) to test for genetic abnormalities that could affect your baby's health.

If you have questions about prenatal diagnostic testing, please ask your physician for additional information.

## (carrier testing

Some birth defects are inherited. A carrier is a person who shows no signs of a particular disorder, but could pass the genetic change on to his or her children.

Parental testing using a blood or saliva sample checks the carrier status for a variety of genetic disorders, including cystic fibrosis, sickle cell anemia, thalassemia, and balanced chromosome rearrangements.



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# prenatal screening



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during pregnancy, your doctor will offer to perform prenatal screening tests to obtain valuable information about the health of your baby.

## (first trimester

First trimester screening is performed between 11 and 14 weeks of pregnancy to determine if the fetus is at increased risk of certain genetic conditions or other congenital abnormalities. The screen includes tests on the mother's blood and an ultrasound scan of the fetus. The blood tests measure certain chemicals in the mother's blood:

- **Pregnancy-associated plasma protein**
- **Human chorionic gonadotropin (hCG)**

The ultrasound scan uses sound waves to visualize nuchal translucency (the amount of fluid underneath the skin at the back of the fetus' neck). Fluid normally accumulates in this tissue, and the ultrasound exam determines if excess fluid is present.

Results from the nuchal translucency screening, the maternal blood test results, and the mother's age are used to calculate the fetus' risk of having Down syndrome, trisomy 18, and other chromosome abnormalities or birth defects.

## (second trimester maternal serum screen

The second trimester screen is performed on the mother's blood between 15 and 20 weeks of pregnancy. This test, called the quad screen, measures the levels of four different chemicals in the mother's blood:

- **Alpha-fetoprotein (AFP)**
- **Unconjugated Estriol**
- **Inhibin A**
- **Human chorionic gonadotropin**

The levels of these substances are used in combination with a woman's age, race, weight and medical history to determine the fetus' overall risk for having a birth defect. These defects include chromosome abnormalities such as Down syndrome or trisomy

18, neural tube defects (incomplete closure of a baby's spine), and other congenital abnormalities.

### General Testing

- Complete Blood Count and Differential
- Rubella Antibody Titer
- ABO-Rh Factor
- Antibody Screen RBC
- Hepatitis B Surface Antigen
- Urinalysis
- Genetic Carrier Testing (if appropriate)

### First Trimester (11-14 weeks)

- Ultrasound (Nuchal translucency)
- PAPP-A
- hCG

### Second Trimester (15-20 weeks) Maternal Serum Screen (Quad Screen)

- AFP
- Estriol
- Inhibin A
- hCG

Prenatal screening is used only to assess risk, not identify a specific genetic condition.