

# Prenatal Genetic Screening Test: For would be parents

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## Introduction

Prenatal genetic testing gives parents-to-be information about whether their fetus has certain genetic disorders. Genetic disorders are caused by changes in a person's genes or chromosomes. Aneuploidy is a condition in which there are missing or extra chromosomes. In a trisomy, there is an extra chromosome. In a monosomy, a chromosome is missing. Inherited disorders are caused by changes in genes called mutations. Inherited disorders include sickle cell disease, cystic fibrosis, Tay-Sachs disease, and many others. In most cases, both parents must carry the same gene to have an affected child.

## Types

There are two types of prenatal tests for genetic disorders:

1. Prenatal screening tests: These tests can tell you the chances that your fetus has an aneuploidy and a few other disorders. This FAQ focuses on these tests.
2. Prenatal diagnostic tests: These tests can tell you whether your fetus actually has certain disorders. These tests are done on cells from the fetus or placenta obtained through amniocentesis or chorionic villus sampling (CVS). FAQ164 [Prenatal Genetic Diagnostic Tests](#) focuses on these tests.

Both screening and diagnostic testing are offered to all pregnant women.

Screening tests can tell you your risk of having a baby with certain disorders. They include carrier screening and prenatal genetic screening tests:

- Carrier screening is done on parents (or those just thinking about becoming parents) using a blood sample or tissue sample swabbed from inside the cheek. These tests are used to find out whether a person carries a gene for certain inherited disorders. Carrier screening can be done before or during pregnancy.
- Prenatal genetic screening tests of the pregnant woman's blood and findings from ultrasound exams can screen the fetus for aneuploidy; defects of the brain and spine called neural tube defects (NTDs); and some defects of the abdomen, heart, and facial features. This FAQ focuses on these tests. They include first-trimester screening, second-trimester screening, combined first- and second-trimester screening, and cell-free DNA testing.

First-trimester screening includes a test of the pregnant woman's blood and an ultrasound exam. Both tests usually are done together between 10 weeks and 13 weeks of pregnancy:

- The blood test measures the level of two substances.
- The ultrasound exam, called a nuchal translucency screening, measures the thickness of a space at the back of the fetus's neck. An abnormal measurement means there is an increased risk that the fetus has Down syndrome (trisomy 21) or another type of aneuploidy. It also is linked to physical defects of the heart, abdominal wall, and skeleton.

Second-trimester screening includes the following tests:

- The "quad" or "quadruple" blood test measures the levels of four different substances in your blood. The quad test screens for Down syndrome, Edwards syndrome (trisomy 18), and NTDs. It is done between 15 weeks and 22 weeks of pregnancy.
- An ultrasound exam done between 18 weeks and 22 weeks of pregnancy checks for major physical defects in the brain and spine, facial features, abdomen, heart, and limbs.

The results from first-and second-trimester tests can be combined in various ways. Combined test results are more accurate than a single test result. If you choose combined screening, keep in mind that final results often are not available until the second trimester.

**Result Interpretation**

Results of blood screening tests for aneuploidy are reported as the level of risk that the disorder might be present:

- A positive screening test result for aneuploidy means that your fetus is at higher risk of having the disorder compared with the general population. It does not mean that your fetus definitely has the disorder.
- A negative result means that your fetus is at lower risk of having the disorder compared with the general population. It does not rule out the possibility that your fetus has the disorder.

Diagnostic testing with CVS or amniocentesis that gives a more definite result is an option for all pregnant women. Your obstetrician or other health care professional, such as a genetic counsellor, will discuss what your screening test results mean and help you decide the next steps.

**How accurate are PGST**

With any type of testing, there is a possibility of false-positive results and false-negative results. A screening test result that shows there is a problem when one does not exist is called a false-positive result. A screening test result that shows there is not a problem when one does exist is called a false-negative result. Your health care professional can give you information about the rates of false-positive and false-negative results for each test.