

A Guide to Prenatal Genetic Testing

There is a lot you can do during your pregnancy to help you have a healthy baby. Taking prenatal vitamins, eating healthy foods, and getting some exercise and enough sleep will help you have a health pregnancy.

The human body is complicated. Even if you do everything “right” during your pregnancy, things don’t always develop normally. Between 3% and 5% of babies have some type of health problem when they are born.

You can be tested during your pregnancy to learn more about your baby’s health before baby is born. Your health care provider will talk about these tests with you and ask you to decide which tests you want. It is your choice whether or not to be tested.

What are the tests?

There are two basic types of tests:

- **Screening tests** predict the *chance*, or odds, that your baby has a certain birth defect.
- **Diagnostic tests** tell you *whether or not* your baby has a certain birth defect.

The table on the next page lists the different tests, along with a timeline a brief description about each one. The following pages include more details about these tests if you would like to read about them before you talk with your doctor.

Screening Tests

Name of Test	When	Description	What it tells you
Nuchal translucency (NT) ultrasound	11 to 13 weeks	Abdominal ultrasound to measure small space behind baby’s neck.	Chances your baby has a chromosome problem.
Integrated screen	11 to 13 weeks and 15 to 22 weeks	NT ultrasound plus 2 separate blood samples.	Chances your baby has Down syndrome, trisomy 18, or spina bifida.
Quad Screen	15 to 22 weeks	1 blood sample	Chances your baby has Down syndrome, trisomy 18, or spina bifida.

Diagnostic Tests

Name of Test	When	Description	What it Tells You
Chorionic villus sampling (CVS)	10 to 13 weeks	Sample of placenta taken vagina or abdomen	<i>Whether or not</i> your baby has chromosome problems and sometimes other inherited diseases
Amniocentesis	15 to 22 weeks	Sample of fluid from around your baby.	<i>Whether or not</i> your baby has chromosome problems, spina bifida, and sometimes other inherited diseases.

Other Tests

Name of Test	When	Description	What it Tells you
Anatomy Ultrasound	18 to 22 weeks	Abdominal ultrasound to check baby's growth and development.	Whether or not abnormalities are suspected and if further testing is needed.