A Guide to Prenatal Genetic Testing

Developing a baby 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 these tests?

Two types of tests:

Screening tests predict the chance that your baby has a certain birth defect

Diagnostic tests tell you whether or not your baby has a certain birth defect

The table below lists different tests, along with a timeline and a brief description of each test.

Screening Tests			
Name of Test	When	Description	What it tells you
Carrier Screening	Pre-pregnancy or any time in pregnancy	Blood test on mother	Looks for abnormalities in mother's DNA that may effect development of the baby for inherited disorders such as Cystic Fibrosis and sometimes others
Nuchal Translucency Ultrasound (NT)	11-13 weeks	Abdominal ultrasound to measure the fat pad on the baby's neck	Chance your baby has a chromosome problem
Integrated screen	11-13 weeks and 15-21 weeks	NT ultrasound plus blood work done at the ultrasound as well as a separate blood test later	Chances your baby has Down syndrome (trisomy 21), Trisomy 18, or a problem with the brain/spinal cord
Non-Invasive Prenatal Testing	Any time after 10 weeks	Blood test on mother looking for fetal DNA **Insurance coverage of this test varies*	Chances your baby has Trisomy 21, Trisomy, 18, Trisomy 13, or problems with the sex chromosomes

Quad Screen	15-22 weeks	1 blood draw	Chances your baby has Trisomy 21, Trisomy 18, or brain/ spinal cord problem
Anatomy Ultrasound	19-22 weeks	Abdominal ultrasound to check baby's growth and development	Whether or not abnormalities are suspected and if further testing is needed

Diagnostic Tests			
Name of Test	When	Description	What it Tells You
Chorionic Villus Sampling (CVS)	10-13 weeks	Sample of placenta taken through the vagina or the abdomen	Whether or not your baby has a chromosome problem and sometimes inherited diseases
Amniocentesis	15-22 weeks	Sample of the fluid from around your baby	Whether or not your baby has chromosome problems, spina bifida, and sometimes other inherited diseases