



Here's What to Expect With Testing & Screening

Today, modern technology makes it possible to know more than ever before about your baby before he or she is born. In addition to routine prenatal appointments, you may wish to consider various types of prenatal testing. Below, you'll find some common tests and their benefits. Different tests can be performed at different stages in your pregnancy, and they are listed in order below.

While there are no tests in pregnancy which provide a 100% guarantee of a normal outcome, chances are in your favor, even with an increased risk assessment, of having a healthy baby.

Carrier Screenings

A carrier screening is done on parents or prospective parents to see whether they carry an abnormal gene for a disorder which could be passed on to their child. A carrier has mild or no symptoms of a disorder, making testing important.

Carrier screening is performed through a blood test. If you are found to be a carrier, your partner is then screened for the disorder. If you are not a carrier, no additional testing is done.

Carrier screening can be done before or during pregnancy. People of certain ethnic groups are more likely to be carriers of different diseases; below you will find more information on the most commonly screened for disorders. All carrier screening has limitations; if you decide to pursue carrier screening you will meet with a genetic counselor to learn about these limitations. Results are usually available within 2-3 weeks.

If you and your partner are both carriers for the same disorder, you may wish to have your baby tested. Ask your healthcare provider or a genetic counselor about your options.

Since these screenings are performed on the parents or prospective parents, they do not need to be re-performed with a subsequent pregnancy.

Prenatal Screening: First Trimester

Non-invasive prenatal testing (NIPT) is a screening test performed any time after 10 weeks 0 days and identifies women whose fetuses are at risk of having an abnormal number of chromosomes. Two of the most common are an extra chromosome ("trisomy") or a missing chromosome ("monosomy"). There are three common trisomy conditions: trisomy 21 (Down Syndrome), trisomy

13 (Patau Syndrome), and trisomy 18 (Edwards Syndrome). A common monosomy is monosomy X (Turner Syndrome). The risk of having an abnormal chromosome count increases with a mother's age.

The **first trimester screen** (FTS) is a prenatal test that is performed between 11 weeks + 6 days and 13 weeks + 6 days of pregnancy and screens for the risk of trisomy 13, 18, and 21. FTS combines two blood markers (PAPP-A and Beta-HGC) and an ultrasound measurement of the thickness of the back of the fetal neck (nuchal translucency). The FTS results provide a revised risk for trisomy 13, 18, and 21 that may be higher or lower than the pre-screen, age-based risk. It does not diagnose the problem definitively.

Follow-up testing: If you have an abnormal NIPT or FTS, you will be referred to a genetic counselor and offered diagnostic testing (such as amniocentesis). The decision to pursue diagnostic testing is a personal decision. The advantage of screening early in your pregnancy is obtaining earlier information about your pregnancy which may allow for more options regarding the pregnancy as well as more time to make those decisions. A genetic counselor will review what your options are for diagnostic testing. If you opt for NIPT or FTS, you will be offered separate screening for neural tube defects in the second trimester.

Prenatal Testing: Second & Third Trimester

Anatomy Ultrasound: 20 weeks

Women usually have at least one ultrasound during their pregnancy. The first ultrasound is usually performed around 20 weeks.

Your baby will be screened for anatomical and placental abnormalities, and abnormalities that increase the risk for aneuploidy. You may also find out the sex of your baby if you wish. If this first "Level 1" ultrasound detects an abnormality, or you have a medical condition associated with increased risk for birth defects, a "level 2" ultrasound can be used to tell you more about the possible risks. Remember that not all abnormalities can be detected by ultrasound. Diagnostic screenings and/or tests will tell you more about your baby.

Gestational Diabetes and Anemia Screening: between 26 to 28 weeks

Gestational diabetes occurs when your body is not able to produce enough insulin during pregnancy. Many hormones secreted by the placenta are insulin inhibitors, so women who are not usually at risk of developing adult-onset (type 2) diabetes can still be affected by gestational diabetes.

If you develop gestational diabetes, diet and exercise changes may be enough to manage this disease. However, women will occasionally need to take insulin shots or diabetes medications to manage symptoms. When untreated, gestational diabetes can lead to high blood pressure and preeclampsia, larger babies, higher risk of c-section, and increases your baby's risk for neonatal (infant) diabetes.

Iron deficiency anemia is common during pregnancy because your body's blood volume doubles and babies need a lot of oxygen. If you develop iron deficiency anemia, your doctor may recommend a multivitamin high in iron, or dietary changes to increase your consumption of meats like chicken, turkey, and beef; or other foods like beans, tofu, spinach, or brown rice.

Tetanus and Diphtheria (Whooping Cough) Vaccine: 28 weeks

The CDC recommends getting the Tdap vaccine between 27-26 weeks. This helps pass immunity to your baby. Getting the vaccine earlier within this timeframe may maximize its effectiveness for your baby. (See [these CDC immunization guidelines for pregnant women](#) for more details).

RhO(D) prescription (for Rh negative blood types): 28 weeks

Rh is a protein found on the surface of red blood cells. About 85% of the population carries the Rh positive protein. If you are Rh negative, and your partner is Rh positive, there is a 50% chance that your baby will be Rh positive. When the baby is positive and the mother is negative, the mother's immune system can be triggered (sensitized) to attack the babies blood cells.

If you are Rh negative, your doctor will prescribe an Rh immunoglobulin shot like RhoGAM. This will prevent Rh sensitization.

Rh sensitization may not affect the first pregnancy as severely, however, if you become pregnant with another Rh-positive fetus, the reaction will be severe and even life threatening to the baby. The best way to prevent serious complications is to prevent sensitization. (Learn more about the [Rh factor and pregnancy](#) on the American Council of Obstetricians and Gynecologists website).

Group Beta Strep test: between 35 to 37 weeks

Group B streptococcus bacteria is commonly found in the gastrointestinal and genital tract. It is unrelated to strep throat. Group B strep is usually harmless in adults, but can be harmful to your baby, with possible effects including pneumonia, meningitis, and blood borne infections or sepsis. It is passed to the baby during labor.

Your provider will perform a Group B strep vaginal and rectal culture between 35-37 weeks gestation. If you are GBS positive, you'll be given antibiotics in labor, or after your bag of water breaks. It's important to know your GBS status (positive or negative) when you are admitted to the hospital for labor and delivery. [Read more about Group B Strep here.](#)



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