

Understanding Prenatal Tests



Prenatal testing can offer you answers and reassurances, but you may have questions about what, exactly, you're being tested for and how the tests work. Can any of the tests reveal genetic abnormalities in your baby? Are any of them dangerous? Are you required to have all the tests done? eP is here to help answer your questions about common prenatal tests that may be recommended for you. Just remember that if you are over age 35, the National Society of Genetic Counselors recommends that you meet with a genetic counselor before undergoing screening tests during pregnancy.

1. Initial Tests

What: You'll be given a long scrip for blood work at your first appointment. Don't be alarmed - all the tests are done with only one needle prick and a few vials of blood. These tests will give your physician information about your blood type, Rh factor (negative or positive blood type), anemia, syphilis, Hepatitis B, HIV, immunity to things like measles and chickenpox, and cystic fibrosis. You'll also get a Pap smear to check for cervical cancer and STDs. Sickle cell anemia testing is ordered for women of African or Mediterranean descent. Urine tests are routinely performed at every prental visit to check the urine for protein (which can signal preeclampsia) and sugar (an indication of gestational diabetes). "The first visit and [set of] blood work are probably most important of all," says Dr. Edmund Funai, chief of obstetrics at Yale-New Haven Hospital.

Who: These tests are routinely done for all women.

When: At your first prenatal visit, you'll be given the scrip and asked to go as soon as possible for the testing. Urine testing will be done at each appointment.

2. Gestational Diabetes

Throughout your pregnancy, you may be sent for additional blood work to test for a variety of conditions, such as gestational diabetes. What: Screening is done with a blood test after you have fasted and then drunk a special glucose drink. Gestational diabetes is an issue for a small percentage of women and can have dangerous consequences for the baby. Dr. Funai reports, "15% test positive and need further testing, but only 30% of these will have diabetes."

When: Routine testing takes place between 24 and 28 weeks, but if you are obese, had a previous baby over 9 pounds, are over 30 or have a family history of diabetes, you may be tested at 12 weeks. If you test positive, you may be sent for a test that requires several blood draws over the space of a few hours.

Who: All women are routinely screened.

3. First Trimester Assessment and Triple or Quad Screening

What: The first trimester assessment is a blood test that screens for trisomy 21 (the chromosomal abnormality known as Down syndrome) and trisomy 18 (another chromosomal abnormality). Triple, quad or AFP testing are blood tests that are done later in pregnancy to further evaluate the risks for these problems, as well as spina bifida. These tests can be done by themselves or in conjunction with an ultrasound, and the latest recommendations show that combining both first trimester and second trimester blood work with an ultrasound gives the best results.

"The AFP/quad tests detect approximately 85% of all cases of trisomy 21 with
only a 7% false positive rate," says Dr.
Stephen Carr, associate professor of obsterrics and gynecology at Brown University.
What is essential to remember about this
test is that it only identifies a risk factor; it
does not diagnose Down syndrome or other
disorders. You don't get a yes or no result
— you get a result that categorizes your
risk, like a 1 in 2,000 or 1 in 400 chance
of having a baby with a genetic disorder.

When: Dr. Carr says, "If a woman wants the earliest possible answer, then I recommend first trimester screening with blood work and ultrasound at 11-14 weeks. If she wants the best, then I recommend first trimester screening with blood work and ultrasound at 11-14 weeks, combined with AFP/quad testing. That combination — the full and integrated screening — yields the best performance and gets results to the woman as early as 16-17 weeks."

Who: Jill Allen Fonda, board member of the National Society of Genetic Counselors, says, "All women are now offered one of these tests, but the current recommendations do not specify which test must be offered," Women over age 35 have a higher risk of Down syndrome and may be encouraged to have the test. If you're considering the test, Dr. Carr recommends that you first evaluate what you are going to do with the information it will provide.

4. Ultrasound

What: Ultrasounds bounce sound waves off your baby's bones and tissue to form an image of her. Ultrasounds are used to determine or confirm due dates, look for multiple babies, check the baby's development, record heartbeats, measure the amount of amniotic fluid and screen for abnormalities.

These tests are believed to be safe for the baby and are usually not uncomfortable — needing to have a full bladder for the test is the biggest complaint by most women. Finding out the baby's sex can be one of the perks of this test, if you want the information. A special ultrasound, called a nuchal translucency test, is used in conjunction with a triple or quad screen (and sometimes first trimester blood work) to evaluate the risk for Down syndrome.

When: The first ultrasound is usually around 18-20 weeks, although some are done earlier to date a pregnancy or check for twins. Later ultrasounds may be done to check positioning of the placenta, to estimate birth weight or during an amniocentesis. Immediate results are often shared, but it may take a week or two for a report to be written.

Who: "Ultrasound is not recommended as a routine procedure by any U.S. professional organization. However, it has become common practice in most of the U.S. and [is] performed [for] almost everyone," points out Dr. Jacques

10. Percutaneous Umbilical Blood Sampling (PUBS)

What: Although the long name makes this procedure sound awfully dire, it is a very simple procedure in which a physician (using ultrasound) guides a needle into the umbilical cord to sample the baby's chromosomes and test the baby for anemia. This test is very quick, with the same sensation as amniocentesis, and offers quick results. The American Pregnancy Association reports the miscarriage risk at 1% or 2% percent.

Who: This test is recommended for women who have had an abnormal ultrasound, who have an inconclusive amnio result or who are exposed to infectious diseases during pregnancy or for babies who have Rh disease.

When: 18-36 weeks, with results in 3 days.

You'll undergo quite a few tests while you're pregnant, so speak up and ask your care provider if you have any questions about them. And try not to stress over them. Remember that they're done to give you and your healthcare team the information you need so you can have the healthiest pregnancy and baby possible.

About the author: Brette McWhorter Sember is the author of Your Practical Pregnancy Planner: Everything You Need to Know about the Financial and Legal Aspects of Preparing for Your New Baby (McGraw-Hill, 2005) and Your Plus-Size Pregnancy: The Ultimate Guide for the Full-Figured Expectant Mom (Barricade Books, 2005).

POLL) Readers Respond

During my pregnancy, I've had the following tests (respondents were asked to check all that apply):

44% Triple screen test

97% Ultrasound

60% Glucose

9% Amniocentesis

8% Chorionic villus sampling



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Abramowicz, director of obstetric/gynecology ultrasound at Rush University Medical Center, High-risk pregnancies may require high-resolution or 3D ultrasounds.

Chorionic Villus Sampling (CVS)

What: A thin tube is inserted through the vagina and cervix (some doctors prefer to insert a thin needle through the abdomen) to take a sample of the chorionic villi, wisps of tissue that attach the placenta to the wall of the uterus. The sample is then tested for Down syndrome and other chromosomal abnormalities (note that CVS cannot test for neural tube defects, but that amniocentesis can). It has a higher risk of miscarriage than amniocentesis (between .5% and 1%, according to the March of Dimes).

When: It is performed at 10 or 11 weeks (a month sooner than amniocentesis), with results in 1-7 days.

Who: This test is used if a first trimester screening indicates a risk of chromosomal problems and/or if a woman is at a higher risk for these and waiting for an amniocentesis is not advised.

Amniocentesis

What: Because it involves inserting a long needle into the belly, the amnio is probably the most feared prenatal test, but it takes approximately a minute, and the needle is thinner than the one used to do a regular blood test. Your physician will use ultrasound to help guide the needle and will take about 1 ounce of amniotic fluid to be tested. Some women feel cramping or a pinch during the procedure.

This test diagnoses Down syndrome, spina bifida and anencephaly conclusively and can also verify the baby's sex. Many women are concerned about the risk of miscarriage after an amnio. That risk has been reported to be 1 in 400, but a recent study showed that when the test was done by a skilled practitioner, the risk was only 1 in 1,000.

When: This test is done at 15-18 weeks with results within a few days or weeks. Who: In general, you should only have an amnio if screening tests have indicated that your risk of an abnormality is higher than the miscarriage risk of the amnio.

7. Non-Stress Test

What: Your baby's heart rate in response to movements he makes is monitored to determine if he is getting enough oxygen and to make sure his nervous system is responding. If your baby is asleep, a noise may be used to wake him up.

When: This is done after the 26th or 28th week.

Who: Non-stress tests are ordered when there is a concern about fetal movement, or if the pregnancy is high risk.

8. Contraction Stress Test

What: The mother is given Pitocin to create uterine contractions. The effect of the contractions on the baby's heart rate is measured and used to determine how the baby will respond to labor contractions.

When: When it is determined by the physician that it is necessary.

Who: This test is usually ordered if a nonstress test has not provided reassuring results.

9. Group B Strep (GBS)

What: Group B streptococcus is a bacterium that, if present in the mother's vagina, can indicate an infection in the baby and cause pre-term labor. The test is done by swabbing the inside of the vagina with a long cotton swab, Dr. Funai says, "30% of all women carry GBS. In many cases, it can be passed to the baby, and if untreated, 1% will get very sick. Screening for GBS and giving antibiotics in labor can reduce 86% of this disease." GBS is easily treated with antibiotics.

When: Between the 35th and 37th week.

Who: This test is routinely done on all women.