Prenatal Testing Information

Should I have Prenatal Testing done?

Although most babies are born in great condition, worrying that your baby may have a problem is common in pregnancy. A variety of tests are available to help detect some of these problems. This worksheet will help you decide if you want any testing done, and if so, what type is right for you. Please review this information and answer the questions at the end **BEFORE** your first visit with the nurse, and bring it with you so we can help make a plan for your pregnancy.

Prenatal tests are used to screen for problems such as chromosome abnormalities (for example Down or Edwards Syndrome), or open neural tube defects (for example spina bifida or anencephaly). Test types fall into two categories: screening tests and diagnostic tests. They can be performed in the first trimester at 10-13 weeks, the second trimester at 15-20 weeks, or in both trimesters. Different types of testing are recommended based on each mother's individual risk.

- Screening tests do not tell you if your baby has a problem, they only tell you if you are above or below the average risk for them. The advantage of screening tests is that they pose little to no risk to you or your baby. The disadvantage is that they don't give you a definitive answer, and they have "false positives" (abnormal test, normal baby) and "false negatives" (normal test, abnormal baby). The chance that an affected baby will have an abnormal screening test (abnormal test, abnormal baby) is called the "detection rate".
- Diagnostic tests are very accurate and will almost always give you definitive yes or no answer. Diagnostic tests are nice because you usually know for sure what's happening, but they carry small risks, sometimes causing miscarriage of a normal baby. So the result of a diagnostic test is more definite than one you get with a screening test, but the risk is slightly higher too. These types of tests are generally recommended if you have an abnormal screening test.

Fetal Anatomy Ultrasound

As a part of your routine prenatal care, your doctor will order a fetal anatomy ultrasound between 18-20 weeks. This ultrasound will be ordered regardless of your decision to have prenatal genetic screening or diagnostic testing.

What is it?

This ultrasound is one of the best ways to see how your baby is developing and offer some reassurance that at this point in pregnancy the baby is developing normally. This is a screening ultrasound, not a diagnostic test. An anatomic Ultrasound will evaluate your baby's growth and major organs, to be sure they have formed properly. It is a special test that gives you a specific glimpse of your growing baby. This test does not check your baby's chromosomes. A prenatal anatomic ultrasound does not detect all physical abnormalities of a baby. During your ultrasound, the baby will be measured from side to side on his/her head, around the head, around the abdomen, and from hip to knee (femur bone). And that's just for starters. The four chambers and blood vessels of the heart will be evaluated, as well as the stomach, kidneys, and bladder of your baby. The internal structures of the baby's brain will be evaluated along with the spine, arms, legs, face and profile. A survey of the baby's anatomy, as well as the placenta location, amniotic fluid levels, and umbilical cord, will help to ensure that the pregnancy is overall developing normally. But if not, it may identify potential problems and thus lead to creating the best possible care plan for you and your baby. The ultrasound will give your doctor a picture of the overall health of your baby and your pregnancy. If you would like to find out the gender of your baby, that too is possible at this time.

What does the mother expect during the ultrasound?

You will lie back on an exam table with your belly exposed, and a sonographer will apply a special gel and move the transducer over your abdomen. As sound waves are emitted from the transducer, they will bounce off "structures" inside your belly and images are formed on the screen. To get the most information from the anatomy of your baby, the sonographer will be obtaining many different views from a lot of different angles. When the sonographer obtains a clear picture, she will freeze-frame the picture for the doctor. The ultrasound can take

Prenatal Testing Information

anywhere from 20 to 50 minutes, depending upon how cooperative your little one wants to be. All electronic devices (including cell phones) must be turned off during the appointment. We don't allow any pictures or recording of the exam, but you can expect to go home with a couple of pictures as well as a CD. The doctor will review the results of the exam following your ultrasound once the sonographer has obtained all of the images and measurements needed.

Screening Blood Tests

Full Integrated Test

This test combines results from two sets of tests, a blood test and ultrasound done at 11-13 weeks, and another blood test done at 15-18 weeks. The results of the ultrasound and both blood tests are analyzed together after the second blood draw. This test screens for Down Syndrome (Trisomy 21), Trisomy 18, and open neural tube defects such (spina bifida for example). This test detects 94% of fetal Down Syndrome, and has a low false positive rate (5%). Results are available by about 16-19 weeks (after your second blood draw). The results will provide you a personalized risk number for each tested condition. For example: Down Syndrome risk is 1 in 1100. Because of its low false positive rate, this test is recommended for patients who are low risk but wish to have more information than that which is provided at the 20 week ultrasound.

Noninvasive prenatal test

A noninvasive prenatal test (NIPT) is a single blood test performed any time at or after 10 weeks. During pregnancy, 3-13% of the DNA in your blood stream is circulating cell free fetal DNA that comes from the placental cells. A NIPT works by evaluating the amount of cell free DNA in your blood. NIPTs screen for Down Syndrome (Trisomy 21), Trisomy 18, and Trisomy 13. This screening test is recommended for patients who are considered to be high risk for fetal chromosome abnormalities. Detection rate for Down Syndrome is reported at 99% in high risk women (those who are age 35+, who have a history of a previous child with a chromosomal abnormality, or those with Robertsonian translocation). Cell free DNA screen does not evaluate the risk of open neural tube defects or any other abnormalities that may be present. This testing is not recommended for low risk patients due to an increased likelihood of false positive results.

Alpha-Fetoprotein test

This test is a single blood test done around 15-22 weeks and assesses only the risk for fetal open neural tube defects. This test may be recommended for high-risk patients who are also having a non-invasive prenatal test.

Look over this table for more information about the pros and cons of each of the above screening tests.

Test	What it tests for	Detection rate	False positive rate	Results available
Full Integrated test	Down Syndrome, Trisomy 18, open neural tube defects	94%	5%	16-20 weeks
Non-invasive prenatal test	Down Syndrome, Trisomy 13, Trisomy 18	99%	0.5% (high risk) Up to 50% (low risk)	10 or more weeks
AFP test	Open neural tube defects	80%	5%	16-22 weeks

Diagnostic Tests

Diagnostic testing options include Chorionic Villus Sampling and Amniocentesis. Please let your physician know if you are interested in either of these testing options.

Prenatal Testing Information

Facts to consider

- Diagnostic tests are more invasive and therefore less safe, but are more accurate
- Screening tests are safer but don't give you a "yes or no" answer
- The genetic conditions being tested for cannot be "fixed", even if we know about them in advance. Having information regarding your baby's genetics allows your provider to optimize your care and your baby's care during pregnancy.
- Prenatal testing is done to gather information, not to find abnormal babies for pregnancy termination. Many parents who would not terminate an abnormal pregnancy have prenatal testing done in order to be prepared for their newborn's special needs.
- Many mental and physical handicaps are not due to chromosome abnormalities or anatomic abnormalities, and are therefore not detectable by blood tests, ultrasounds, or amnio/CVS.

Circle your age-based risk

Age at delivery	Down Syndrome risk (Trisomy 21)	Total risk for Trisomy 21, Trisomy 18, and Trisomy 13
33	1/625	1/345
34	1/500	1/277
35	1/385	1/204
36	1/303	1/167
37	1/227	1/130
38	1/175	1/103
39	1/137	1/81
40	1/106	1/63
41	1/81	1/50
42	1/64	1/39
43	1/50	1/30
44	1/38	1/24
45	1/30	1/19

Adapted from Schreinmachers, DM, Cross, PK, Hook, EB. Rates of trisomies 21,18,13 and other chromosome abnormalities in about 20,000 prenatal studies compared with estimated rates in live births. Hum Genet 1982; 61:318. In UpToDate.