

Prenatal Testing Options

While most babies in the United States are born healthy, between three and four percent are born with a birth defect, and two to three percent will have developmental delays. All parents face these risks, but some factors, including maternal age, family history and ancestry can increase these risks.

A variety of prenatal screens and diagnostic tests are available to parents. Choosing the right test or screening can be difficult and confusing. The following summary is intended to provide some information to help in the decision-making process. A discussion with a doctor or genetic counselor may also be helpful. Remember, no screen or diagnostic test can address all birth defects or retardation and these tests are directed at specific problems.

What is screened for?

Some of the birth defects that may be addressed in routine testing options include Down syndrome, a chromosome abnormality associated with mild to moderate mental disability, a rarer chromosome problem called trisomy 18, associated with severe developmental disabilities, and spina bifida, a defect in the spinal cord.

What kinds of tests are available?

Two types of tests are available to all pregnant women: screening tests and diagnostic tests.

A screening test is intended to identify pregnancies that face an unexpected 'high' risk of having a specific birth defect. If a screening test is abnormal, it means that an increased risk exists and a diagnostic test will be offered. Most individuals who receive a positive or abnormal screen have healthy babies. A diagnostic test provides definitive testing for the condition. Most diagnostic tests are over to 99% accurate.

Screening Tests:

One of the best screens available is a complete family history. A family history includes information on ancestry, health, presence or absence of birth defects, retardation, autism, miscarriages, infertility, and other major medical problems for all extended family members. Things in a family history can influence the type of tests that are offered to you and the accuracy of those tests.

First trimester screening is done after 11 weeks of pregnancy. It is designed to assess the risk for two of the age-related chromosomal conditions, Down syndrome and trisomy 18. There are three ways of doing the first trimester screen.

1. The first trimester "Firstscreen" involves an ultrasound to measure the back of the baby's neck and a blood test. It is done between 11 and 14 weeks of pregnancy. The results take about one week. This screen can alert us to about 83-85% of pregnancies affected with Down syndrome. If the screen is abnormal, then an amniocentesis is offered for diagnostic testing.
2. The first trimester sequential screen involves the neck ultrasound and blood test described above. A preliminary result is available in one week. If the results of the preliminary screen show a "high" risk, then a diagnostic test is suggested. If this preliminary result indicates a Down syndrome risk that is "low", then a second blood test

is done. This second blood test result is then combined with the results of the first blood test to give a final result. This option alerts us to over 90% of pregnancies affected with Down syndrome. It also allows us to detect most babies affected with spina bifida.

3. The first trimester integrated screen is similar to the sequential screen; however, no preliminary result is available. The blood test and ultrasound are done between 11-14 weeks and then a second blood test is done at 16 weeks to provide the final result. This screen can alert us to over 90% of pregnancies affected with Down syndrome and will also detect most babies affected with spina bifida. If the screen is abnormal for Down syndrome or trisomy 18, then an amniocentesis is offered for diagnostic testing.

-Second trimester serum screening is done in pregnancies that are beyond 15 weeks and have not had a first trimester screen. It involves a blood test that measures proteins produced by your baby. It will alert us to over 80% of women who carry a baby with Down syndrome. It will also alert us to most babies with spina bifida.

DIAGNOSTIC TESTS:

-Diagnostic tests provide definitive testing for specific birth defects. Diagnostic tests are relatively safe, however, they are associated with a small increased risk of miscarriage.

CVS is the earliest diagnostic test available. This is usually done between 10-12 weeks of pregnancy. This test allows evaluation of the chromosomes. It is greater than 99% accurate in addressing the risk for chromosomal imbalances such as Down syndrome.

Amniocentesis is the most common diagnostic test. This test is done after 15 weeks. It also allows evaluation of the chromosomes and is greater than 99% accurate. It appears to have a slightly lower risk for miscarriage compared to the CVS.

Making Decisions

For parents who want to know with certainty if a specific problem is present or absent, a diagnostic test is best. For someone who wants to clarify their risks before deciding about a diagnostic test, a screening test may be more appropriate. For some parents, neither screening nor diagnostic tests is appropriate.

During pregnancy, patients are asked to make many health care decisions. In many cases there isn't a clear "good" or "bad" choice. The best decision is one that feels right for the patient and circumstances. It may be helpful to meet with a physician or a genetic counselor to review your risk factors, discuss your options and define your personal goals for prenatal testing.