

Screening Tests for Birth Defects

Screening tests can help assess the risk of having a baby with a birth defect. Some women choose to have screening tests depending on their age, medical history, previous pregnancies, family history or ethnic background. Most women, however, choose to have screening for reassurance. This handout provides a brief overview of screening tests.

Birth Defects

Almost all children in the United States are born healthy. Out of 100 newborns, only two to three have major birth defects. For about 70% of babies born with birth defects, the cause is not known. In other cases, birth defects are inherited through genetics or caused by the mother being exposed to harmful agents or medications.

Most birth defects occur during the first three months of pregnancy. Some can be found before birth with screening tests. The most common birth defects found through screening tests include:

- *Neural tube defects*: incomplete closure of the fetal spine can result in spina bifida or anencephaly (absence of part of the skull)
- *Abdominal wall defects*: failure of the abdominal wall to close during development
- *Heart defects*: mild to severe abnormalities of heart shape or function
- *Down syndrome*: mental retardation, abnormal facial features, and other medical problems occur as a result of an extra chromosome 21 (also called trisomy 21)
- *Trisomy 13 and 18*: there is an extra chromosome 13 or 18, either of which causes severe mental retardation and severe birth defects

Types of Screening Tests

Screening tests are easy to perform and pose little risk to you or your fetus.

Screening tests do not diagnose a birth defect, but they tell you if you are at higher risk of having a child with a birth defect than the average person. If the risk is high, you may choose to proceed with more invasive diagnostic testing. The following screening tests are available and routinely performed.

First Trimester Screen: This is a noninvasive screening test that estimates the chances that a woman may have a baby with Down syndrome, trisomy 13 or trisomy 18. The test is usually done between 11 4/7 and 13 6/7 weeks gestation. This screen involves a special ultrasound of the baby and a maternal blood test. First trimester screening is 85% accurate at detecting Down syndrome and 95% for trisomies 13 and 18.

Alpha-fetoprotein (AFP): This is a maternal blood test for neural tube defects, such as spina bifida and anencephaly. It is done between 15 and 20 weeks. AFP detects neural tube defects 80% of the time. AFP is included in the quad screen and can also be done alone after a first trimester screen.

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UW Health Ob-Gyn Clinics

20 S. Park St. Clinic*
20 S. Park St., Suite 307
20 S. Park St., Suite 506
Madison, WI 53715
(608) 287-2830

Arboretum Ob-Gyn Resident
Physician Clinic
1102 S. Park St.
Madison, WI 53715
(608) 287-5898

Union Corners Clinic*
2402 Winnebago St.
Madison, WI 53704
(608) 242-6840

West Clinic*
451 Junction Rd.
Madison, WI 53717
(608) 265-7601

*Location includes
Midwifery Clinic

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Quad Screen: This is a maternal blood test that measures four substances (AFP, estriol, human chorionic gonadotropin and inhibin-A), which are altered in Down syndrome, trisomies 13 and 18, and neural tube defects. The quad screen is done between 15 and 20 weeks and detects Down syndrome 80% of the time, trisomy 18 60-70% of the time and neural tube defects 80% of the time.

Anatomy Ultrasound: All pregnant women are offered a comprehensive ultrasound between 20 and 22 weeks. This detailed survey of the fetus' organs and features can detect some birth defects.

Other Screening Tests

Several other screening tests are available to detect the following diseases:

- Cystic fibrosis is a severe respiratory and digestive disease most common among Caucasians.
- Spinal muscular atrophy is a disorder that results in progressive muscle weakness and paralysis. It affects males and females.
- Thalassemia is a type of anemia more common in people of Mediterranean, Middle Eastern, African or Asian descent.
- Sickle cell anemia is another type of anemia common to people of African descent.
- Tay-Sachs is a disorder of fat metabolism that causes severe mental retardation, blindness and seizures and is more common in Ashkenazi Jews and French Canadians.

For Higher Risk Patients

Your baby may be at higher risk for Down syndrome and other chromosome abnormalities if you:

- Are age 35 or older at the time of delivery
- Have a personal or family history of a chromosome problem
- Have abnormal results on the first trimester screen or quad screen
- Have an ultrasound with abnormal findings

Options for higher risk patients include cell free fetal DNA testing, CVS (chorionic villus sampling), and amniocentesis. If you are considering any of these tests, you will need to be seen by a genetic counselor at either Meriter or St. Mary's.

Cell Free Fetal DNA Testing – (Also referred to as “non-invasive prenatal testing or NIPT”) This is a blood test that measures the baby's DNA (genetic material) from the mother's blood to determine if the baby might have a chromosome problem. It is about 99% accurate, so if the test is abnormal, it is very likely, but not certain, that the baby has the condition. Further testing can be done to determine with even more certainty if the baby has the condition or not. Not all insurance covers the cost of this test, so there may be an out-of-pocket cost. This test can be done after 10 weeks gestation.

Chorionic Villus Sampling (CVS) – this is a diagnostic prenatal test that examines placental tissue to determine if a baby has a chromosomal abnormality. The test is usually done between 11 4/7 and 13 6/7 weeks gestation.

Amniocentesis – This is a diagnostic prenatal test that examines cells found in the amniotic fluid to determine if a baby has a chromosomal abnormality. The test is typically done after the 16th week of pregnancy.

In Summary

Your provider will discuss with you the available screening tests and what the results mean. If the results raise concerns about your pregnancy, additional diagnostic tests may be offered. Talk to your provider about which screenings are right for you.