



## Screening for Birth Defects

Screening tests are done during pregnancy to assess the risk of certain birth defects.

### **FIRST TRIMESTER SCREENING:**

First trimester screening combines the results of an ultrasound and blood tests, which are done between 11-13 weeks of pregnancy. This screening detects signs of Down syndrome, Trisomy-18 and possible heart defects.

The ultrasound test is called nuchal translucency screening. This test measures the thickness at the back of the neck of the fetus. An increase in this measurement may be a sign of Down syndrome, Trisomy-18 or heart defects.

Two blood tests are performed, pregnancy associated plasma protein – A (PAPP-A) and free beta human chorionic gonadotropin (hCG).

The combined results of the nuchal translucency screening and the blood tests show if the fetus might have Down syndrome, Trisomy-18 or a heart defect. If a nuchal translucency is increased but the combined screening does not show an increased risk of Down syndrome or Trisomy-18, a detailed examination of the fetal heart is done later in pregnancy at 20 weeks.

### **SECOND TRIMESTER SCREENING:**

In the second trimester, maternal blood testing is used for screening for Down syndrome, neural tube defects, Trisomy-18 and abdominal wall defects. These blood tests include alpha-fetoprotein (AFP), Estriol, Human chorionic gonadotropin (hCG) and Inhibin-A. The test is called a quad screen. This screening test is performed between 15-20 weeks of pregnancy.

Because first trimester screening is done too early to check for neural tube defects, maternal blood is drawn for AFP between 15-20 weeks to be tested for this problem.

If first trimester screening has been done, only AFP and not a full quad screen will be checked in the second trimester.

A detailed ultrasound examination of the baby between 19-22 weeks evaluates the baby's anatomy. While ultrasound is an excellent and safe noninvasive screening tool, it will only pick up 50% of defects, leaving 50% undetected that will only be found at birth.

If a screening test result shows an increased risk for having a baby with a certain defect, further tests can help diagnose the problem. In most cases, the baby is healthy even if there is an abnormal screening test result, false/positive. A specialized ultrasound or amniocentesis may be discussed in the case of an abnormal screening test result.



## Diagnostic Tests:

### Chorionic Villus Sampling and Amniocentesis

**CHORIONIC VILLUS SAMPLING (CVS):**

This procedure involves taking a sample of placental cells to test for genetic abnormalities. This sampling of cells is done either through the cervix or through the abdomen at 10-12 weeks. A perinatology specialist performs this test. An ultrasound is required prior to seeing the perinatologist for this appointment.

CVS cannot detect neural tube defects, i.e. anencephaly or spina bifida, and an alpha-fetoprotein test done at 16-18 weeks is recommended for neural tube defect screening. Results of the CVS are generally known in 7-10 days. The risks of CVS include miscarriages (approximately 1%), infection, possible limb defects, mouth and jaw defects, bleeding or cramping.

**AMNIOCENTESIS:**

Amniocentesis is performed between 15-20 weeks of pregnancy. A needle is placed through the abdomen and into the uterus to remove a small amount of amniotic fluid under ultrasound guidance. This amniotic fluid is sent to the lab and cells are grown on a special culture. The cells are then evaluated for chromosomal abnormalities such as Down syndrome or Trisomy-18. The alpha-fetoprotein level in the amniotic fluid can also be tested to determine if the fetus has a neural tube defect.

Complications from amniocentesis are uncommon; however, they can include mild cramping, vaginal bleeding, infection and amniotic fluid leakage. There is a slight chance of miscarriage, 0.5% as a result of the amniocentesis.

Mother's Age	Risk of Down Syndrome	Risk of Any Chromosomal Disorder
20	1/1,667	1/526
25	1/1,250	1/476
30	1/952	1/385
35	1/378	1/192
36	1/289	1/156
37	1/224	1/127
38	1/173	1/102
39	1/136	1/83
40	1/106	1/66
41	1/82	1/53
42	1/63	1/42
43	1/49	1/33
44	1/38	1/26
45	1/30	1/21