What are CVS and amniocentesis?

CVS and amniocentesis are tests used to diagnose conditions such as Down syndrome, trisomy 18, and trisomy 13. CVS is done by taking a small sample of cells from the placenta. Amniocentesis is done by taking a small sample of the amniotic fluid. CVS is done between 10-14 weeks of pregnancy and amniocentesis is done after 15 weeks of pregnancy. Both of these tests have a risk for pregnancy complications or miscarriage. The risk for miscarriage as a result of CVS is about 1 in 200 pregnancies (0.5%) and the risk for miscarriage as a result of amniocentesis is 1 in 300 to 1 in 500 pregnancies (0.2%-0.3%).

Is first trimester screening covered by insurance?

First trimester screening is covered by many insurance companies. The ultrasound and blood work are billed separately. If you have questions about billing or insurance for the blood test, you can reach the billing department for NTD Labs at 1-888-683-5227. Financial counselors are available at UNC to discuss the cost of ultrasound.

How do I schedule first trimester screening?

Tell your healthcare provider you are interested in first trimester screening. Remember, it must be done early in pregnancy so tell your healthcare provider as soon as possible.

Will I be offered other screening tests in my pregnancy?

You will be offered a screening test in the second trimester to see if the chance for spina bifida or another type of neural tube defect (NTD) is increased. Screening for NTDS can be done on a sample of your blood. The blood test is called AFP, or alpha-fetoprotein. A targeted ultrasound in the second trimester can also detect most neural tube defects.

If you do not have first trimester screening, you will be offered a different screening test in the second trimester that screens for spina bifida, trisomy 18, and Down syndrome. This test is called the maternal serum screen (also called the quad screen). It is done on a sample of the mother’s blood. If you have first trimester screening, you will not need a maternal serum screen, because you will have already had screening for Down syndrome and trisomy 18.

What if I have more questions?

If you have more questions or are uncertain if you wish to have first trimester screening, you can request an appointment to meet with a genetic counselor early in your pregnancy to review all of your screening and testing options.

If you have questions about first trimester screening or other available services, please call Reproductive Genetics at: 919-966-2229
What is first trimester screening?
First trimester screening is an optional test offered to all pregnant women. Results can tell you if there is an increased chance for Down syndrome, trisomy 18, or trisomy 13 in the pregnancy.

What is Down syndrome?
Down syndrome is the most common genetic condition seen in newborns. It is sometimes called trisomy 21. Babies with Down syndrome have intellectual disabilities and differences in their facial features. Babies with Down syndrome may also have problems with their heart and other organs. Down syndrome typically happens by chance and does not run in families.

What are trisomy 18 and trisomy 13?
Trisomy 18 and trisomy 13 are other genetic conditions sometimes called Edwards syndrome and Patau syndrome. Both of these conditions are less common than Down syndrome. Babies with trisomy 18 or trisomy 13 usually have severe birth defects and often do not live very long after birth. Many pregnancies with trisomy 18 or trisomy 13 result in miscarriage or stillbirth. Like Down syndrome, trisomy 18 and trisomy 13 typically happen by chance and do not run in families.

How is first trimester screening done?
First trimester screening involves a blood test and ultrasound. Usually, the blood is collected through a finger stick. There are two ways the screen can be done:

- **Traditional method (11-14 weeks):** You will meet with a genetic counselor before the ultrasound to discuss the screening test, fill out the paperwork, and have your blood collected. Your genetic counselor will call you with your result about five days later.

- **Instant risk assessment (IRA) method (blood test > 9 weeks, ultrasound 11-14 weeks):** Your blood test will be done by your healthcare provider at least one week before the ultrasound. Results are usually available immediately after the ultrasound. The doctor will discuss the results with you after your ultrasound.

How will the results of my screening come back?
The results will either be reported as “within range” or “increased risk”.

What if my test results are “within range”?
A “within range” result means that the chance of Down syndrome is less than ~1 in 300 (~0.3%). For trisomy 18 and trisomy 13, a “within range” result means the chance of both conditions is less than 1 in 150 (0.67%). This does not completely rule-out the possibility of these conditions in the pregnancy. However, 85% of pregnancies with Down syndrome and 90% of pregnancies with trisomy 18 or trisomy 13 will be detected by this screening test.

What if my test results show an “increased risk”?
An “increased risk” result means that the chance for having a baby with Down syndrome, trisomy 18, or trisomy 13 is above our cut-off. It is important to remember that this result does not mean your pregnancy has any of these conditions. If your test results show an increased risk, you will be offered further testing such as chorionic villus sampling (CVS), amniocentesis, or ultrasound.

Why is an ultrasound done?
The ultrasound measures the amount of fluid found at the back of the baby’s neck. This is called the nuchal translucency (NT). An increased amount of fluid can mean a higher chance of Down syndrome, trisomy 18, trisomy 13, and other problems. However, extra fluid can also be seen in babies with no health problems. The ultrasound also measures the length of the baby to make sure that the test is being done at the right time.

Why is a blood test done?
The blood test measures proteins in the mother's blood which are made by the growing pregnancy. If levels of these proteins are different than the average pregnancy, it can mean an increased chance for Down syndrome, trisomy 18, or trisomy 13.