Wilmington Maternal-Fetal Medicine

PRENATAL SCREENING/TESTING OPTIONS

All women are offered screening for chromosomal abnormalities in their pregnancy. Some women want this information and some women do not. The below information is to help you understand screening options. Insurance may not cover all of these tests. You may need to contact your insurance to discuss coverage and cost. You can decide which, if any, are right for you.

SCREENING TESTS: Non-invasive. Provides a higher or lower risk evaluation and does not make a diagnosis in pregnancy.

Ultrasound/NT: Most insurances cover this testing. In the first trimester, nuchal translucency (NT) is a neck measurement that can be performed to assess risk for Down syndrome. In the second trimester, an anatomy ultrasound (level II, targeted ultrasound) can be performed to screen for birth defects such as spina bifida.

Maternal serum (AFP/Sequential) screening: Most insurances cover this testing. This is a non-invasive blood test that screens for spina bifida, Down syndrome, or trisomy 18. Sequential screening is a two part test that involves a first trimester ultrasound measurement for NT and two blood draws from the mother; one in the first trimester and one in the second trimester, with results one week after the second blood draw. AFP/sequential screening can identify 80-90% of babies with Down syndrome and trisomy 18 and 80% of babies with spina bifida. It does not tell gender.

NIPS (cell free fetal DNA, cffDNA, NIPT): Most insurances cover this test only for women at increased risk for Down syndrome and other chromosome conditions. This is a non-invasive blood test that looks at circulating cell-free fetal DNA from a mother's blood. This test screens for Down syndrome, trisomy 18, trisomy 13 and extra or missing sex chromosomes. NIPS provides up to a 99% detection for Down syndrome. Detection for trisomy 18 and trisomy 13 may be slightly lower. It does not screen for spina bifida.

DIAGNOSTIC TESTS: Invasive with a small risk to a pregnancy; used to diagnose chromosome conditions in a baby. Most insurances cover only for women at increased risk for Down syndrome and/or other chromosome conditions.

Chorionic villus sampling (CVS): Performed in the first trimester. A small sample is taken from the placenta to study a baby's chromosomes. Used to diagnose Down syndrome, trisomy 18, trisomy 13 and sex chromosome conditions. It does not test for spina bifida.

Amniocentesis (Amnio): Performed in the second trimester. A small sample of fluid is taken from the amniotic sac inside of the uterus. Used to diagnose Down syndrome, trisomy 18, trisomy 13 and sex chromosome conditions. It can also test for spina bifida.