WHAT IS NONINVASIVE PRENATAL TESTING?
Noninvasive Prenatal Testing (NIPT) is a relatively new and advanced screening test for chromosome conditions. Currently, this test screens pregnancy for several chromosome conditions including Down syndrome (also known as trisomy 21), trisomy 18, or trisomy 13. Babies with Down syndrome have distinctive physical features with mild to moderate intellectual disability, and may also have other birth defects such as heart problems. Trisomy 18 and Trisomy 13 are less common than Down syndrome and are serious conditions that include birth defects, severe disability, and shortened lifespan. NIPT determines if one of these conditions is likely to be present or not in a pregnancy. This testing can be performed as early as the 10th week of pregnancy.

WHO IS OFFERED NIPT?
NIPT is offered when there is an increased suspicion for one of the chromosome conditions included in the test. The most common reason why women are offered NIPT is due to maternal age. Women 35 years or older at the time of delivery have an increased chance to have a pregnancy with a chromosome condition and are routinely offered options for prenatal testing that also include chorionic villus sampling (CVS) or amniocentesis. Other women who are offered NIPT include those who have had a prior pregnancy with a chromosome condition or women with either an ultrasound finding during the pregnancy or another blood screening that increases the chance for a chromosome condition regardless of the women’s age.

HOW IS NIPT DONE?
NIPT involves a blood sample that is taken from the mother’s arm. During pregnancy, there are small pieces of the baby’s genetic maternal (fetal DNA) that have passed through the placenta and into the mother’s blood. DNA is the genetic material that makes up chromosomes. We know that Down syndrome (trisomy 21), trisomy 18 and trisomy 13 are due to an extra chromosome 21, 18 or 13, respectively. NIPT measures the amount of genetic material from chromosomes 21, 18, and 13. If extra genetic material is detected, it can indicate an increased chance for the baby to have a chromosome condition. Results for this test come back either positive (increased risk) or negative (decreased risk).

ARE THE RESULTS OF NIPT ACCURATE?
The accuracy of NIPT results vary between the different laboratories and chromosome conditions. NIPT identifies greater than 98% of pregnancies with Down syndrome and trisomy 18 and 80 - 92% of cases of Trisomy 13. Since NIPT is still considered to be a screening test, a diagnostic test is recommended to confirm a positive result of the NIPT. A negative NIPT result rules out each condition in greater than 99% of cases. NIPT is most accurate for singleton pregnancies. In twin pregnancies in which only one baby has a trisomy, NIPT cannot tell which baby is affected in the event of a positive result.

WHAT OTHER TESTING SHOULD BE OFFERED AFTER NIPT?
In the event of a positive result for a chromosomal condition from NIPT, a woman may opt for diagnostic testing to get more information about her pregnancy. The two diagnostic testing options during pregnancy are chorionic villus sampling (CVS) and amniocentesis. CVS is performed during the first trimester between the 11th and the 12th week and involves sampling a small piece of the placenta. Amniocentesis is usually done after the 15th week of pregnancy and involves taking a sample of amniotic fluid from around the baby. As these are invasive tests, there is a risk for miscarriage. However, CVS and amniocentesis are the most accurate prenatal tests available for chromosome abnormalities and may be offered to any woman who desires diagnostic prenatal testing.

Since NIPT does not test for openings along a baby’s spine (neural tube defects/spina bifida), a blood screening called maternal serum AFP screening for neural tube defects should be offered between 15 to 18 weeks in the pregnancy. In addition, a second trimester ultrasound at 18-20 weeks gestation is usually performed to check the growth and development of the baby. Ultrasound screens for other birth defects that NIPT cannot detect such as heart defects or cleft lip that are part of any woman’s risk when she becomes pregnant. Approximately 3-5% of all babies born have some unexpected finding or birth defect.

IS NIPT THE RIGHT TEST FOR ME?
The decision to have a prenatal diagnostic test or advanced screening test is a personal one. A woman should think about the benefit of knowing the chromosome information about the baby during the pregnancy. It may be helpful to discuss these options with your husband/partner, family, physician, and/or genetic counselor. Prenatal genetic counselors help families understand how genetic conditions are occur and what prenatal testing options are available to determine if a baby has a birth defect or genetic condition present. A genetic counseling session is recommended as part of the noninvasive prenatal testing process.