Palmetto Health USC
MEDICAL GROUP
Department of Obstetrics and Gynecology
USC Genetic Counseling

NONINVASIVE PRENATAL SCREENING

WHAT IS NONINVASIVE PRENATAL SCREENING?
Noninvasive Prenatal Screening (NIPS) is a relatively new and advanced screening option for chromosome conditions. Currently, this test screens a pregnancy for several chromosome conditions including Down syndrome (also known as trisomy 21), trisomy 18, and 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. NIPS determines if one of these condition 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 NIPS?
NIPS 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 NIPS 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 NIPS 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 woman's age.

HOW IS NIPS DONE?
NIPS involves a blood sample that is taken from the mother's arm. During pregnancy, a small amount of placental DNA is present in the maternal 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. NIPS 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 NIPS ACCURATE?
The accuracy of NIPS results vary between the different laboratories and chromosome conditions. NIPS identifies greater than 98% of pregnancies with Down syndrome and trisomy 18 and 80 - 92% of cases of Trisomy 13. Since NIPS is still considered to be a screening test, a diagnostic test is recommended to confirm a positive result from NIPS. A negative NIPS result rules out each condition in more than 99% of cases. NIPS is most accurate for singleton pregnancies. In twin pregnancies, in which only one baby has a trisomy, NIPS cannot tell which baby is affected in the event of a positive result.

WHAT OTHER TESTING SHOULD BE OFFERED AFTER NIPS?
In the event of a positive result for a chromosomal condition from NIPS, 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 10th and the 13th 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. These are safe procedures, but do carry some risk for miscarriage. 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 NIPS does not screen for openings along a baby's spine (neural tube defects/spina bifida), a blood screen called maternal serum AFP (MSAFP) should be offered between 15 and 18 weeks of pregnancy to determine the risk for a baby to be affected with spina bifida. In addition, a second trimester ultrasound at 18-22 weeks gestation is usually performed to check the growth and development of the baby. Ultrasound screens for other birth defects, such as heart defects or cleft lip, which NIPS cannot test for. Birth defects are part of any woman's risk when she becomes pregnant, as 3-5% of all babies born have some unexpected finding or birth defect.

IS NIPS 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 screening process.

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