

Non-Invasive Prenatal Testing or NIPT is a screening test for certain chromosomal abnormalities in the fetus. This test is performed on a sample of the mother's blood any time after 10 weeks of gestation. The maternal blood sample analyzes cell free DNA of the placenta which is identical to the baby in the majority of circumstances. This test cannot detect all genetic changes that could cause health problems but it does screen for some of the most common genetic conditions which include Down Syndrome, Trisomy 18, Trisomy 13, Monosomy X (Turner's Syndrome), Sex chromosomal abnormalities, Triploids and, if you desire, the gender of the baby. A normal test is reassuring but is not a guarantee of a healthy pregnancy or baby. This test is a screening test, not a diagnostic test, so if the result indicates your baby is at an increased risk for a chromosomal abnormality, then you will be referred for further testing. Further diagnostic testing is highly accurate and is either chorionic villus testing (biopsy of the placenta) or amniocentesis (removal of some fluid surrounding the baby).

Genetic Carrier Screening This is a maternal blood test to determine if the mother is a carrier for certain genetic conditions. This test does NOT screen the baby. A carrier for a genetic disorder is someone who has one gene for the condition being tested and is not affected. A carrier has no symptoms and no idea she carries a gene unless previously tested. An affected person has two genes and symptoms of the genetic disorder. ACOG recommends offering hemoglobinopathy testing at the initial prenatal visit if no prior testing results are available (H14 or larger). If you pursue testing and are found to be a carrier you may wish to consider genetic carrier testing for your partner, consult with your Obstetrician or pursue genetic counseling.

Patients mistakenly think that if they have no family history of any genetic disorders they are unlikely to be affected. A study of Cystic Fibrosis patients (CF is a genetic disorder that causes frequent lung infections and a shorten life span) revealed that 80% of children diagnosed after birth had no family history.

There are several genetic panels you can choose from depending on how many genetic disorders you want to screen for. The smallest panel is 3 genetic disorders and the largest is 421 genetic disorders, but you may choose from 3, 4, 14, 27, 106, 274 or 421 different genetic disorders. Talk to your Obstetrician about which panel is best for you.

Current genetic testing can detect the majority of carriers but no test can detect 100% of all carriers. If your test is negative there is a small risk you could still be a carrier.

The decision to pursue screening is up to you. If you would like to speak with a Genetic Counselor about these tests you can log in to support@natera.com or call 650-489-2124 for Panorama testing or 650-489-1303 for Carrier Screening. This will allow you to make an educated decision on your genetic screening choices.

_____ I DESIRE Non Invasive Prenatal Testing NIPT/Cell Free DNA

_____ I DESIRE Carrier Screening for the following number of genetic disorders

3_____ 4_____ 14_____ 27_____ 106_____

274_____ 421_____

_____ I DECLINE Non-Invasive Prenatal Testing NIPT/Cell Free DNA

_____ I DECLINE Carrier Screening

Patient's Signature_____

Date _____