
PRENATAL TESTING Informational Handout

This handout summarizes the types of prenatal genetic testing and birth defect testing available during your pregnancy. These tests are optional, but the consequences of failure to identify a birth defect prior to the birth of the baby can be serious. Please review this handout carefully and ask about anything you do not understand or would like more information about.

Genetic Carrier Blood Test Panel for Recessive Mutations (Inheritest)

This is a blood test on either the mother, the father, or both parents, that checks for DNA mutations. A person can be normal and still carry a mutation for a recessive genetic condition. If both parents carry the same recessive mutation (or trait), then there is a 25% chance the baby can be born with a serious genetic disease, caused by inheriting the recessive trait (mutation) from each parent. Cystic Fibrosis is the most common mutation, but mutation panels can test for more than 100 conditions.

NIPT (Non Invasive Prenatal Test – sometimes called cell-free DNA or cfDNA, MaterniT 21+)

This is a blood test on the pregnant woman that can be done starting about 9 weeks gestational age that can detect and analyze fetal DNA fragments that circulate in the mother's blood. The fetal DNA is tested for gender (male or female); chromosome abnormalities (including Down Syndrome-also known as Trisomy 21 (T21), Trisomy 18, Trisomy 13, Turner Syndrome-missing an X chromosome, and other conditions including microdeletions (missing fragments of DNA). We urge patients to investigate cost information with their insurance providers.

Nuchal Translucency Ultrasound (NT scan)

About 11-13 weeks gestation, an ultrasound is done and the width of an area on the back of the neck of the fetus is measured. A normal width is less than 3 mm. A width of 3 mm or greater could be due to a birth defect and additional testing is advised.

Nuchal Translucency (NT) Blood Test

The California Prenatal Screening Program (PNS) provides screening for a few birth defects (T21, T18, and spina bifida mainly). The program consists of 1 or 2 blood tests. The first one is the NT blood test. The second one is the afp blood test (see below). The NT blood test is usually waived if the NIPT is done (see above). Abnormal results of this blood test usually warrant additional testing. *see (www.cdph.ca.gov/Programs/CFH/DGDS/Pages/pns/default.aspx)

AFP Blood Test (alpha-fetoprotein blood test)

This blood test is usually done about 16 weeks and can be done by itself or as the second part of the California Prenatal Screening Program. One condition this test can help identify is a rare birth defect known as spina bifida (abnormal development of the brain or spinal cord).

Anatomy Ultrasound (anatomy scan)

This ultrasound is usually done at 18-20 weeks. We examine the baby head to toe, looking at the brain, heart, spine, limbs, and internal organs and perform standard measurements of the baby.