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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 biological parents, that checks for DNA mutations. A person can be normal yet still carry a mutation for a recessive genetic condition (heterozygous). If both parents carry the same recessive mutation (or trait), then there is a 25% chance (1 in 4) the baby could be born with a serious genetic disease, caused by inheriting the recessive trait (mutation) from each parent (homozygous). Cystic Fibrosis is the most common mutation, but mutation panels can test for more than 500 conditions, most very rare.

NIPT (Non Invasive Prenatal Test – sometimes called cell-free DNA or cfDNA, MaterniT 21 PLUS)
This is a blood test on the pregnant woman that can be done starting 9 weeks gestational age and 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 or T21), Trisomy 18, Trisomy 13, Turner Syndrome (missing an X chromosome or 45X0), and other conditions including microdeletions (missing fragments of DNA). We urge patients to investigate cost information with their insurance providers. The State of California offers a less comprehensive version of this test than the one we advise.

Nuchal Translucency Ultrasound (NT scan)
About 11-13 weeks gestation, an ultrasound is done and the width of an area on the fetal neck is measured. Normal is less than 3 mm. A width of 3 mm or greater could be due to a birth defect and additional testing is advised, usually referral to a MFM (maternal-fetal medicine) specialist.

Nuchal Translucency (NT) Blood Test
With the widespread use of NIPT, the NT blood test is rarely done or needed. If NIPT is not done, this blood test can help determine if a higher than expected risk of T21 or T18 is present. If so, then NIPT might be a useful confirmatory test. A fetus can have a normal NT scan and normal NT blood test and potentially still be born with T21. Not all cases of T21 can be identified by screening, which is one of the reasons we offer NIPT to all pregnant patients.

AFP Blood Test (alpha-fetoprotein blood test)
This blood test is usually done about 16 weeks and can be done by itself or as 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.