Genetic Testing Options

A list of your Testing Options during Pregnancy

Non-Invasive Screening Tests *First Line Blood Testing*	**Timing – (Gestational Weeks)**
MaterniT 21 Plus- Non-invasive prenatal testing (NIPT) Risk factors for abnormal chromosome testing includes: 35 years old and older, sono markers, positive carrier screening, previous affected child, parent with translocation. Screens for Trisomy 21 (>99% accurate, 0.1% false positive) Trisomy 18 (99% accurate, 0.4% false positive) Trisomy 13 (91% accurate, 0.3% false positive) Sex chromosome anomalies (96% accurate, 0.3% false positive) Can do with twins	9 weeks and above
Nuchal Translucency Combines ultrasound of fetal neck skin thickness with blood test for levels of certain markers 90% detection rate of Trisomies 21 & 18 5% false positive Can do with twins	11 weeks and 3 days – 13 and 6 days
Tetra Screen Blood test for certain markers 75-78% detection Trisomy 21 73% Trisomy 18 up to 80% neural tube defects False positive 3 – 5%	15 weeks – 21 weeks and 6 days
Invasive Diagnostic Tests *Performed by MFM specialist if abnormal initial screen or patient request based on prior history – Not performed at P&M*	
Chorionic Villus Sampling Full chromosome analysis of placenta cells. 1:300 risk pregnancy loss	10 weeks – 13 weeks and 6 days.
Amniocentesis Full chromosome analysis of fetal cells from amniotic fluid. 1:300 risk pregnancy loss	16 weeks and above

Additional Non-Invasive Testing Options	**Timing – (Gestational Weeks)**
Inheritest CF/SMA Panel - Genetic Carrier Screening Partner needs carrier testing if positive **Cystic Fibrosis (CF) Carrier Screening** 1:24-94 carrier rate (varies by ethnicity) Screens for most common, but not all, mutations. **Spinal Muscular Atrophy (SMA) Carrier Screening** 1:35-117 carrier rate (varies by ethnicity) affects 1:6000-10,000 babies annually Respiratory failure and respiratory infection are causes of death Muscles degenerate and atrophy due to death of nerve cells in spinal cord	1st visit ideal, can be done at any time during or before pregnancy.
Inheritest Core Panel - Genetic Carrier Screening Partner needs carrier testing if positive Includes Cystic Fibrosis (CF) and Spinal Muscular Atrophy (SMA) Carrier Screening described above, as well as Fragile X syndrome. **Fragile X Carrier Screening** 1: 259 carrier rate for females A leading inherited cause of intellectual and developmental disabilities Fragile X carrier screening is recommended for women with a family history of fragile X-related disorders or intellectual disability suggestive of fragile X syndrome	1st visit ideal, can be done at any time during or before pregnancy.
Inheritest Ashkenazi Jewish Panel - Genetic Carrier Screening Partner needs carrier testing if positive Screens for more than 40 disorders specific to individuals of Ashkenazi Jewish descent – including CF, SMA, Fragile X, and Tay-Sachs disease	1st visit ideal, can be done at any time during or before pregnancy.
AFP (alpha fetoprotein)- non-genetic Measures risk of open neural tube defects Detects 80% of ONTD 1 – 3% false positive rate	15 weeks – 23 weeks and 6 days

I have personally explained the above testing options to the patient or the patient's designated decision maker.

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Provider Signature Date

The above testing options have been explained to me.

_____ _____

Patient Signature Date

Patients are encouraged to get personalized cost estimates, insurance coverage information and more from LabCorp by going to www.integratedgenetics.com

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