SCREENING AND DIAGNOSTIC TESTING OF FETAL CHROMOSOMES

Non-Invasive Screening

Nuchal Translucency (NT) Screening: For many years, we have been offering a first trimester test which calculates the risk of the fetus having extra chromosomes, called trisomies. This test uses a combination of the mother's age, hormones that come from the fetal placenta, and a sonogram measuring the pocket of fluid at the base of the baby's skull. This screening test has a 90-93% detection rate of Down Syndrome (Trisomy 21) and a 5% false positive rate.

Cell-Free Fetal DNA Screening: Tests are now available that can isolate fetal DNA from the developing placenta. These cells can be normally be found in maternal blood after 10 weeks gestation. These newer tests have a Down Syndrome detection rate of 99.1%. The American College of Ob-Gyn issued a statement in 2015 recommending that cell-free fetal DNA screening continue to be offered **ONLY** to high risk women; because of this statement, our office will not pre-certify these tests for low-risk women. Low risk women must be prepared to pay cash if they choose this testing method.

Depending on which test, which insurance carrier you have, and whether you have met your deductible or have co-insurance, the cost even for high risk women can vary tremendously. Our practice is obliged by contracts we have with managed care companies to use in-network labs whenever possible.

Test for High-Risk Women only:

MaterniT21 PLUS (Lab: Sequenom: 877-821-7266, option 2). In-network with Oxford, United, Aetna, Cigna and Multiplan, so those insurers will decide how much their subscribers will have to pay. If insurance will not be billed, Cash Price $995. Five trisomies, seven microdeletions and sex chromosomes examined.

Tests for High and Low-Risk Women:

Informed Pregnancy Screen (Lab: Counsyl: 888-268-6795) In-network with Oxford, United, Cigna and Aetna.; can be sent through Enzo Lab for BCBS. Cash price if not covered by insurance $349; additional $250 for microdeletions.

Harmony (Lab**:** Ariosa: 855-927-4672) Cash price $130 ($117 if pay within 30days of billing)

Panorama (Lab: Natera; sent through Bioreference 800-229-5227) Cost will vary depending on insurance contracts.

VisibiliT: Sequenom's low-risk test; looks for Trisomies 21 & 18 and reports sex. Cash price $140.

This field is evolving rapidly; please inform yourself and investigate your insurance options if you are interested in pursuing these tests.

Invasive Diagnostic Testing

Both Amniocentesis and chorionic villus sampling (CVS) are still available to high risk women or to confirm a positive result on non-invasive testing. These invasive tests carry a small risk of miscarriage, but can be used for a greater variety of genetic diagnoses, for example, if both parents carry the gene for a certain disease.

Blood for NT screening should be drawn between 9wks 3d & 11wks: _____

Blood for Fetal DNA test for HIGH-RISK only drawn after 10wks: _____

Sonogram for Nuchal Translucency should be bet. 11 ½ -13 ½wks: _____

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