TESTING FOR CHROMOSOMAL ABNORMALITIES

As a woman ages, her risk for chromosomal abnormalities such as Down Syndrome increases. These abnormalities can, however occur in a woman of any age. Several tests are available to assess a woman's risk. Screening tests are non-invasive and do not involve any risk to the fetus. There are both false positive and false negative results with these tests. "Positive" results need to be confirmed by a diagnostic test. Diagnostic tests are invasive procedures that carry small inherent risks of miscarriage. These tests provide a definitive answer as to whether there are abnormal chromosomes.

Screening Tests

1. **Nuchal Translucency screening (NT)**

Recommended by the American College of Obstetrics and Gynecology (ACOG). This screening is performed between 11 and 14 weeks. Blood tests are combined with a sonographic measurement of the thickness at the back of the fetal neck, or nuchal fold, to detect > 90% of fetuses with Down Syndrome and 98% of fetuses with Trisomy 13 &18 with a false positive rate of 5%. This test is available to women of any age.

2. **AFP –** The blood test for alpha feot-protein (AFP) is recommended at 15-16 weeks to assess for Open Neural Tube defects such as Spina Bifida. Often this test is combined with the NT.

3. **Non Invasive Blood Chromosomal Screening (MateriT 21, Panorama, Harmony)**

Recommended by ACOG for patients 35 years and older, or with a previous chromosomal abnormality or family history. Blood test that screens for Trisomy 13, 18 & 21 as early as 9 weeks gestation. If abnormal, further diagnostic testing would be recommended.

If you desire this test, please check with your insurance company for coverage.

Diagnostic Tests

1. **CVS (Chorionic Villus Sampling)**

This test is performed between 10 and 13 weeks. Cells are removed from the placenta either transvaginally or transabdominally and the chromosomes are analyzed. The risk of miscarriage is 1 in 100. This test does not evaluate Open Neural Tube defect risks (AFP) which is done at 15 – 16 weeks.

2. **Amniocentesis**

Starting at 15 weeks, an amniocentesis can be performed by inserting a needle under ultrasound guidance through the mother's abdomen and uterus into the amniotic sac to remove a small amount of fluid from around the fetus. Chromosomes are then analyzed from this fluid as well as AFP levels to evaluate for the Open Neural Tube defects. The risk of miscarriage while originally thought to be .3 - .5% now appears to be much lower and is quoted in some reviews to be 1 in 1600 or .06% based on current data.

I have received a copy of the Testing for Chromosomal Abnormalities and have been informed of my options.

Name_____

Signature_____

Date_____