Prenatal Testing Options for Your Pregnancy
Patient Decision Aid

NON-INVASIVE PRENATAL TESTING (NIPT) – NIPT is a newer blood test that looks at fetal DNA found in the mother's blood. This test helps determine if your fetus is at risk for Down syndrome (trisomy 21) and two other chromosome conditions called trisomy 18 and trisomy 13. It may also test for fetal sex and differences in the number of sex chromosomes. NIPT is more accurate than traditional screening methods. Results take approximately 8-10 days. Only 1 in 1,000 women will receive a false positive result. False negative results are rare. Invasive diagnostic testing (CVS or amniocentesis – see description below) is needed to confirm whether or not the fetus is affected.

NUCHAL TRANSLUCENCY (NT) SCREENING – The nuchal translucency is a pocket of fluid behind the fetal neck. It can be seen by ultrasound in the first trimester of pregnancy (11-14 weeks). NT screening measures the size of this pocket. Fetuses with larger NT's have a higher risk for Down syndrome and other genetic conditions. NT screening alone can detect 64% - 70% of Down syndrome cases[2]. Invasive diagnostic testing (CVS or amniocentesis – see description below) is needed to confirm whether or not the fetus is affected.

INTEGRATED SCREENING – Integrated screening involves NT screening (see above) and two blood draws, one at 10-14 weeks and the second at 15-21 weeks. Integrated screening can detect up to 92% of fetuses with Down syndrome (trisomy 21), 90% of fetuses with trisomy 18, and 80% of fetuses with open spina bifida (a type of neural tube defect). Integrated screening can also be done without including the NT measurement but at a lower detection rate. Results are not reported until the second trimester when all parts of the test have been reviewed. One in twenty women will receive a false positive result. Amniocentesis (see description below) is needed to confirm whether or not the fetus is affected.

Second trimester screening (Quad) – Second trimester screening involves a blood draw at 15-21 weeks gestation. This testing can detect up to 80% of fetuses with Down syndrome (trisomy 21), 80% of fetuses with trisomy 18, and 80% of fetuses with open spina bifida (a type of neural tube defect). Results are reported 1-2 weeks after the blood draw. One in twenty women will receive a false positive result. Amniocentesis (see description below) is needed to confirm whether or not the fetus is affected.

FETAL ANATOMY ULTRASOUND – This ultrasound is generally done between 18-20 weeks of pregnancy. It uses sound waves to create images of the fetus. Ultrasound can help identify problems with the growth and development of the baby. Some findings seen on ultrasound may increase the chance that the fetus could have Down syndrome (trisomy 21) or another chromosome condition. However, ultrasound alone cannot diagnose chromosome conditions. There is no known risk to the pregnancy.

CHORIONIC VILLUS SAMPLING (CVS) – This test can be done between 10-13 weeks of pregnancy. A small sample of chorionic villi, cells that form the placenta, are collected and grown in the laboratory. Then, the chromosomes in those cells are studied. This procedure can be done through the cervix using a catheter or through the belly using a needle. CVS is >99% accurate for the detection of chromosome conditions. It has a risk of miscarriage of <1/100 (1%)[3]. CVS cannot detect neural tube defects like spina bifida.

AMNIOCENTESIS-This test is generally done between 16-22 weeks of pregnancy. Using ultrasound guidance, a needle is inserted through the belly into the amniotic sac. A small amount of the amniotic fluid around the fetus is taken out. Cells from the fetus that are in the amniotic fluid are grown in the laboratory. Then the chromosomes in those cells are studied. Amniocentesis is >99.8% accurate for the detection of chromosome conditions. It has a risk of miscarriage of 1/300 to 1/500[4]. Amniocentesis can also detect neural tube defects like spina bifida by measuring alpha-fetoprotein (AFP) in the amniotic fluid.

REFERENCES

1. Unless otherwise specified, all detection rates and false positive rates are taken from the following: Integrated Genetics. "Maternal Serum Screening – Product Offerings."

2. ACOG Practice Bulletin No. 77, January 2007. Screening for fetal chromosomal abnormalities. *American College of Obstetricians and Gynecologists*. *Obstet Gynecol*. 2007 Jan; 109(1):217-27.

3. Chorionic villus sampling. Retrieved from http://www.mayoclinic.com/health/chorionic-villus-sampling/MY00154/DSECTION=risks.

4. Amniocentesis. Retrieved from http://www.mayoclinic.com/health/amniocentesis/MY00155/DSECTION=risks

Integrated Screening	Harmony Prenatal Test	Amniocentesis
SCREENING TEST ▸ Provides a risk assessment for Down syndrome and trisomy 18	**SCREENING TEST** ▸ Provides a risk assessment for Down syndrome, trisomy 18, and trisomy 13. ▸ **Optional**: Can provide a risk assessment for other chromosomal conditions, such as Turner syndrome and Klinefelter syndrome. Fetal sex determination is also possible.	**DIAGNOSTIC TEST** ▸ Determines whether or not the fetus has a chromosome condition ▸ Tests for a wider variety of chromosome conditions than either screening test
DETECTION RATE: ▸ 92% of fetuses with Down syndrome ▸ 90% of fetuses with trisomy 18 ▸ 5% (1 in 20) chance of receiving a “false positive” result	**DETECTION RATE:** ▸ >99% of fetuses with Down syndrome ▸ >98% of fetuses with trisomy 18 ▸ 8 of 10 fetuses with trisomy 13 ▸ 0.1% (1/1,000) chance of receiving a “false positive” result	**DETECTION RATE:** ▸ >99% of fetuses with Down syndrome ▸ >99% of fetuses with trisomy 18 ▸ >99% of fetuses with trisomy 13 and other chromosome conditions ▸ Minimal risk for “false positive” result
WHAT IS INVOLVED? ▸ Blood draw at 10-14 weeks AND ▸ Ultrasound to measure fetal nuchal translucency (NT) at 11-14 weeks AND ▸ Second blood draw at 15-20 weeks	**WHAT IS INVOLVED?** ▸ One blood draw anytime after 10 weeks of pregnancy	**WHAT IS INVOLVED?** ▸ Removal of a small amount of amniotic fluid via a needle inserted in to the abdomen ▸ 16-22 weeks of pregnancy ▸ The risk for pregnancy complications, including miscarriage, is 1/300 to 1/500.
WHEN DO I GET RESULTS? ▸ Combined results in the second trimester	**WHEN DO I GET RESULTS?** ▸ 8-10 days after blood draw	**WHEN DO I GET RESULTS?** ▸ 1-2 weeks following the procedure