

FURTHER INFORMATION FOR WOMEN WHO CONSIDER HAVING AN INVASIVE TEST

Basic information on invasive tests is provided below. This office provides amniocentesis services in conjunction with genetic counseling. We can provide you the names of several physicians in the region who perform chorionic villus sampling.

WHAT IS CHORIONIC VILLUS SAMPLING (CVS) AT 11-14 WEEKS?

The test is offered at 10-12 weeks gestation. This involves the examination of chorionic villi (placental cells). Both the baby and the placenta (afterbirth) develop from the same cell. That is why the cells of the placenta can be used to check the chromosomes of the baby.

HOW IS CVS DONE?

CVS can be done from the transabdominal approach or through the cervix. Which technique is used depends primarily upon the particular physician performing the procedure and the location of the placenta.

TRANS ABDOMINAL CVS

Local anesthetic is given. A fine needle is then passed into the uterus through the mother's abdomen and a sample of cells is taken. The needle is watched carefully with an ultrasound scan to ensure that it does not harm the fetus. The whole procedure takes 2-3 minutes.

TRANS CERVICAL "CVS"

The doctor inserts a thin tube, or a catheter through the cervix into the placenta. When gentle suction is applied, a chorionic villus sample is withdrawn.

NEED TO REPEAT THE TEST...

In about 1% of cases, the invasive test will need to be repeated because the cells will not grow in the laboratory or the results are inconclusive.

WHAT ARE THE RISKS ASSOCIATED WITH THE TEST?

The risk of miscarriage due to CVS is 1% (1 in 100), approximately twice that of amniocentesis.

AMNIOCENTESIS IS 15 WEEKS OR LATER

Amniocentesis is another invasive test which involves passing a thin needle into the uterus in order to take some fluid from around the fetus. This test gives the same information as CVS but it is carried out at 15 weeks and the results are available 2 weeks later. The risk of miscarriage is 1 in 200 to 1 in 400, about one half that of CVS.



WHAT MAKES THE MATERNAL FETAL IMAGING DIVISION OF HUDSON VALLEY RADIOLOGY UNIQUE?

HVRA is in partnership with NTD Laboratories who have the proprietary license for the free beta HCG maternal biochemical analyte which confers statistically significant reduction in false positives.

- 1. Consultation** with board certified physicians who are subspecialty fellowship trained in high risk Maternal Fetal Imaging.
- 2. Accreditation:** We are the first providers of 11-14 week First Trimester Screening (accredited by The Fetal Medicine Foundation, London, England) and one of the few private practice maternal-fetal imaging programs who have achieved and maintained accreditation in obstetrical ultrasound and fetal echocardiography by the prestigious American Institute of Ultrasound in Medicine (AIUM). Evidence-based publication in a peer review journal has demonstrated statistically significant superiority in first trimester screening programs that are required to adhere to the rigorous quality assurance program of the Fetal Medicine Foundation.
- 3. Detailed protocols** to assess your fetus not only for structural malformations but for the most extensive list of genetic ultrasound "markers": a partial list includes—middle bone of the pinky finger, widened space between first and second toes of the foot, ear and nasal bone length, and our unique assessment of cardiovascular Down Syndrome markers.

4. Universal Fetal Cardiac Assessment: Approximately 60% of Down Syndrome fetuses and approximately 80% of trisomy 18 fetuses have structural heart diseases. **The detection of structural heart disease therefore is a critical requirement of ultrasound risk assessment for chromosomal abnormalities. We are the only private practice providers of genetic obstetrical ultrasound studies whose physicians also have the dedicated expertise in fetal echocardiography (detailed evaluation of the fetal heart).**

5. Quantitative Down Syndrome Risk Assessment: The end result of your exam will be a number that represents the risk of Down Syndrome or Trisomy 18 that is unique to your fetus. Each patient has their own unique emotional balance sheet that weighs the risk of a chromosomal abnormal baby with the risks and benefits of knowledge gained from an amniocentesis. The purpose of this consultation is to provide the information allowing you to make a decision that is best for you (Do I want or need an amniocentesis?). The appropriateness of nasal bone ossification as a Down syndrome marker requires subspecialty accreditation. Normal nasal bone ossification for gestational age increases first trimester screening Down syndrome detection (sensitivity) from 85 to 95%.

6. Sophisticated Imaging Tools: If a fetal structural abnormality is found, we have the most sophisticated imaging technology including Fetal MR (magnetic resonance imaging) and 3D Ultrasound, to fully analyze the problem so you can be counseled appropriately.

OUR LOCATIONS

HVRA of Inter-County

955 Yonkers Ave, Room 14
Yonkers, NY 10704
P: 914-776-7700

HVRA of Westchester

115 Main St
Tuckahoe, NY 10707
P: 914-793-1234

HVRA of Bronxdale

2016 Bronxdale Ave, Ste 102
Bronx, NY 10462
P: 718-960-9033

Mid Rockland Imaging

18 Squadron Blvd
New City, NY 10956
P: 845-634-9729

New Paltz Imaging

3 Cherry Hill Rd, Ste 1
New Paltz, NY 12561
P: 845-255-9700

MATERNAL FETAL IMAGING SERVICES

UNIVERSAL FETAL
CARDIAC ULTRASOUND—
AT THE HEART OF
NEWBORN WELL-BEING

NON-INVASIVE SCREENING FOR FETAL
STRUCTURAL MALFORMATIONS AND
CHROMOSOMAL ABNORMALITIES: FIRST
TRIMESTER SCREENING AT 11-14 WEEKS

BRIEF OVERVIEW:

- The vast majority of babies are born normal.
- All women, whatever their age, have a small risk of delivering a baby with a physical and/or mental handicap.
- In some cases the handicap is due to a chromosome abnormality such as Down syndrome (Trisomy 21 or Trisomy 18).
- First Trimester Screening (FTS) gives an estimate of the risk for Down Syndrome. To know for sure whether or not the fetus has a chromosome abnormality, an invasive test is needed (chorionic villus sampling or amniocentesis). However, invasive tests carry a small risk of causing miscarriage.

WHAT IS DOWN SYNDROME?

Down Syndrome is a chromosomal disorder that causes mental retardation and birth defects which may involve the heart and/or digestive tract. Down Syndrome affects approximately one in every 800 babies.

RISKS FOR DOWNS IN RELATION TO MATERNAL AGE

The table shows how the chance of having a baby with Down Syndrome increases with age. In the beginning of pregnancy, the risk that the fetus has a chromosome abnormality is higher than at birth because many affected fetuses die naturally during pregnancy.

Maternal age related risks of having a live born child with Down Syndrome and the risk of carrying a fetus with Downs at 12 weeks of gestation.

RISK OF TRISOMY 21

AGE	AT BIRTH	AT 12 WEEKS
20	1 in 1526	1 in 1018
25	1 in 1351	1 in 901
30	1 in 894	1 in 596
32	1 in 658	1 in 439
34	1 in 445	1 in 297
36	1 in 280	1 in 187
38	1 in 167	1 in 112
40	1 in 96	1 in 64
42	1 in 55	1 in 36
44	1 in 30	1 in 20



WHAT IS TRISOMY 18?

Trisomy 18 is a more severe chromosomal disorder which causes profound mental retardation and multiple birth defects. Few Trisomy 18 babies live longer than one year, although some do survive childhood. Like Down Syndrome, the chance to have a pregnancy with Trisomy 18 increases with increased maternal age.

WHAT IS FIRST TRIMESTER SCREENING: THE 11-14 WEEK SCAN

First Trimester Screening consists of a combination of an ultrasound exam and a maternal blood test performed between 11-13 weeks and six days (the crown rump length: 45-84 millimeters) of pregnancy. The ultrasound exam assesses gestational age and looks for major physical defects. Then, the amount of fluid accumulation behind the neck of the baby, called nuchal translucency (NT) is measured. A maternal blood specimen is obtained and analyzed for two chemicals called free beta-hCG (human chorionic gonadotropin) and PAPP-A (pregnancy associated plasma protein-A), which are normally found in the blood of all pregnant women. The results of the ultrasound exam are combined with the results of the blood test to estimate a specific risk level for Down Syndrome and Trisomy 18.

WHAT DOES FIRST TRIMESTER SCREENING TELL ME AND MY DOCTOR?

First Trimester Screening can provide helpful information about the possibility of chromosomal disorders and structural malformations. The test alerts you and your doctor that your baby may be at increased risk for one of these disorders. The 11-14 week scan can detect some but not all physical defects. A follow-up ultrasound study at 18-22 weeks with a Cardiovascular Genetic/Level II Ultrasound is recommended to optimize noninvasive ultrasound risk assessment for chromosomal abnormalities and structural malformations. If the ultrasound measurement of the nuchal translucency is enlarged (greater than 2.5 mm) there is an increased risk of structural (anatomic) abnormalities even if chromosome studies (amniocentesis or CVS) are normal. Under such circumstances, we recommend a Level 2 fetal ultrasound and a dedicated fetal cardiac ultrasound (fetal echocardiography) at 18-22 weeks.

Please remember that if your test result does not fall within the normal range, it only means that further testing (CVS or amniocentesis) may be indicated. Since First Trimester Screening is a screening test, and not a diagnostic test, an abnormal result does not necessarily mean that there is a problem with your baby. Depending on the estimated risk, you may be happy to continue without further testing.

WHAT HAPPENS IF FIRST TRIMESTER SCREENING SHOWS THAT I AM AT INCREASED RISK?

If the First Trimester Screening test indicates an increased risk for chromosome abnormalities, your doctor will refer you for genetic counseling to explain the test results and your options for further testing. These options may include an invasive test such as chorionic villus sampling (CVS) or amniocentesis. The advantage of an invasive test is that it is diagnostic— you get a diagnosis, or definitive answer. The disadvantage is the risk of miscarriage (see discussion later in this overview). Alternatives to invasive testing include additional screening tests which provide a risk estimate for Down Syndrome but are not diagnostic: Modified sequential screen maternal blood test at 15 weeks; Cardiovascular Genetic/Level II Ultrasound, which detects approximately 98% of Down fetuses if both are normal.

HOW DOES FIRST TRIMESTER SCREENING COMPARE TO OTHER WAYS OF DETERMINING IF MY FETUS IS AT RISK FOR A CHROMOSOMAL ABNORMALITY OR A STRUCTURAL MALFORMATION?

Currently, advanced maternal age (AMA, >35 yoa) and an abnormal maternal blood second trimester screen test (15-16 weeks) are the methods used to determine risk and select which patients would benefit from amniocentesis. First Trimester Screening (FTS) has several advantages over these approaches. FTS is more sensitive. For example, FTS will detect 90% of Down Syndrome in comparison to second trimester screen, 60-70% and AMA 20-30%. FTS has significantly less false positive and that means you are far less likely to be exposed to the risks of an invasive procedure (amniocentesis) following a positive FTS in comparison to advanced maternal age (AMA) or a positive second trimester screen.

Our program of First Trimester Screening and 18-22 week Cardiovascular Genetic/Level II Ultrasound is the most sensitive and specific way to non-invasively optimize risk assessment for chromosomal abnormalities and structural malformations.

HOW ACCURATE IS FIRST TRIMESTER SCREENING?

90% of babies with Down Syndrome and 75% of Trisomy 18 babies will have a positive First Trimester Screening test. In addition, patients carrying babies with other chromosomal disorders or birth defects may have a positive First Trimester Screening. However, a negative First Trimester Screening does not eliminate the possibility that the baby may have Down Syndrome, Trisomy 18 or other chromosomal abnormalities, nor does the test eliminate the possibility of birth defects, mental retardation and other disorders not detectable by prenatal screening. Therefore we recommend that all women have an 18-22 week Cardiovascular Genetic/Level II ultrasound scan. In the majority of cases, no increased risk is found and patients can be reassured that it is unlikely that their baby had Down Syndrome or Trisomy 18.

Please Remember: The decision to have First Trimester Screening is your personal decision. If these tests show that you are at increased risk, it does not mean that your baby has a problem, only that further evaluation of your pregnancy is indicated. In addition, please speak to your doctor about second trimester maternal serum AFP (alpha-fetoprotein) to screen for neural tube defects (NTD's).