

the maternal fetal imaging division of hudson valley radiology associates introduces

The CardioVascular Genetic / Level 2 Obstetrical Ultrasound Exam: 90 % Down Syndrome detection sensitivity

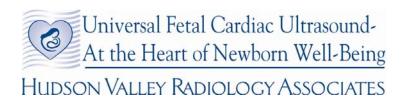


Mid-Rockland Imaging

18 Squadron Blvd New City, NY 10956 845-634-9729 **HVRA** of New Paltz

279 Main Street, Ste. 103 New Paltz, NY 12561 845-255-9700 **HVRA** of Westchester

115 Main Street Tuckahoe, NY 10707 914-793-1234



UPDATE GENETIC ULTRASOUND PROTOCOLS AND DOWN SYNDROME DETECTION SENSITIVITY

Dear Colleague,

This communication serves to introduce the <u>Cardiovascular Genetic/Level II Ultrasound</u> exam – a detailed maternal-fetal ultrasound study with advanced cardiovascular protocols achieving a Down syndrome risk detection rate (sensitivity) of at minimum 90%. When combined with normal first, modified sequential or second trimester quad screen, Down syndrome risk detection may be as high as 99%.

The following represent the new ultrasound views:

- cardiac Doppler (CPT 76827) for tricuspid valve regurgitation.
- cardiovascular color flow mapping (CPT 93325) for
 - interventricular septal defect
 - tricuspid valve regurgitation
 - aberrant right subclavian artery
 - aberrant umbilical and IVC venous drainage patterns and agenesis ductus venosus.

The following references are the evidence-based literature supporting updated cardiovascular ultrasound protocols to complete and optimize Down syndrome risk detection.

Role of Second Trimester Genetic Sonography After Down Syndrome Screening. Aagaard-Tillery. Obstetrics and Gynecology. Vol. 114, #6. December 2009. pp.1189-1196.

First and Second Trimester Ultrasound Screening for Down Syndrome – Can They be Combined? Bromley. Advanced Sonography Symposium in OB-GYN. Harvard Medical School Department of Continuing Education. October 2010.

Genetic Sonography: The Historical and Clinical Role of Fetal Echocardiography. Devore. Ultrasound Obstetrics Gynecology. 2010; 35: 509-521.

Trisomy 21: 91% Detection Rate Using Second Trimester Ultrasound Markers. Devore. Ultrasound Obstetrics Gynecology. 2000: 16: 133-141.

Aberrant Right Subclavian Artery: Marker for Chromosomal Abnormality. Borenstein. Ultrasound Obstetrics Gynecology 2010; 46: 548-552.

Umbilical Vein Anomaly in Fetuses with Down Syndrome. Achiron. Ultrasound Obstetrics Gynecology 2010; 297-301.

Prenatal Diagnosis of Ductus Venosus Agenesis and Its Association with Cytogenetics/Congenital Anomalies. Prenatal Diagnosis. 2002; 995-1000.

The target population for cardiovascular genetic ultrasound would be those patients in whom you desire to complete and optimize Down syndrome risk assignment. A partial list might include the following patient populations:

- Late registrant and/or unscreened pregnancy
- Advanced maternal age with or without screening
- Increased aneuploidy risk on screening studies without karyotyping

As with all our mid second trimester ultrasound anatomy studies, patients are invited to participate in our Pregnancy Outcome Quality Assurance Program.

New prescription pads will be provided. What was previously labeled as Genetic/Level II Detailed Obstetrical Ultrasound will now be called Cardiovascular Genetic/Level II Detailed Obstetrical Ultrasound.

Please contact me if you have any questions or comments, either by cell phone (914) 391-0109 or by email danjcohen@optonline.net.

Sincerely,

Daniel J. Cohen, MD

DJC:ec



FAST FACTS ON FETAL CONGENITAL HEART DISEASE (CHD)

- Incidence, 1:200 1:300 pregnancies with the profound majority having no risk factors
- CHD is the most common, the most serious and nationally the most frequently missed of all fetal malformations
- Current detailed/level II accreditation guidelines have not improved the 15-30% national detection rate of fetal CHD. This failure acknowledges that "it is not the mere performance of 'outflow tract' imaging but the detailed knowledge of fetal cardiac pathology, its recognition and the interpretive expertise that is necessary to substantially increase detection rate of CHD."
- CHD is the malformation most responsible for infant morbidity and mortality accounting for greater than one-third of infant deaths related to congenital malformation.
- Without a prenatal diagnosis, even severe forms of congenital heart disease commonly go undetected until after discharge to home leading to avoidable morbidity and mortality.
- 20-55% of infants with CHD are not diagnosed until after hospital discharge. Most obstructive left heart lesions (such as a ortic coarctation) are not diagnosed at birth or at six weeks.
- Aortic coarctation is one of the three undiagnosed conditions (the others are hypoplastic left heart and interrupted arch) most likely to lead to death soon after discharge from hospital.

Prenatal Detection of Congenital Heart Disease in Southern Nevada, The Need for Universal Cardiac Evaluation. J. Ultrasound in Medicine, 26:1715-1719.

Prenatal Screening of Major Congenital Heart Disease. J. Ultrasound in Medicine, 28: 889-899.