What is the role of fetal echocardiography (2D 76825, cardiovascular color flow mapping 93325) as performed in conjunction with detailed fetal anatomy scan (CPT 76811) in comparison to AIUM's new outflow tract protocols?

I. **Fetal echocardiography as performed and interpreted by MDs subspecialty trained and accredited in the recognition and diagnosis of fetal cardiac disease remains the evidence-based "gold standard" to achieve 90-100% detection of congenital heart disease (CHD) and to complete and optimize risk assessment for aneuploidy and nonchromosomal syndromes.**

It is not the mere performance of "outflow tract" imaging but the detailed knowledge of fetal cardiac pathology, its recognition, and the interpretative expertise that is necessary to substantially increase detection rate of CHD.

• Acherman. Prenatal detection of congenital heart disease in southern Nevada. The need for universal fetal cardiac evaluation. *J Ultrasound Med 2007*. 26:1715-1719.
• Yagel. Congenital heart defect: Natural course and in utero development. *Circulation 1997*; 96:550-555.
• Behtiyar and Copel. Improving detection of fetal cardiac of fetal cardiac anomalies – a fetal echocardiogram for every fetus? *J Ultrasound Med 2007*. 26:1639-1641.
• Stümpflen. Effect of detailed fetal echocardiography as a part of prenatal ultrasound screening in the detection of congenital heart disease. *Lancet 1996*. 348: 854-857.

Summary –

CPT 76805 – Nondetailed Obsterical Ultrasound- four chamber and proximal outflow tract views

_ less than 40-50% CHD detection.
_ no evidence based Down syndrome (DS) detection

CPT 76811 – Detailed 'level II' Obstetrical Ultrasound

_ less than 60-70% CHD detection.
_ 50% DS detection.

HVRA's Cardio Vascular Genetic Detailed L2 Obstetrical Ultrasound.
CPT 76811, 76825, 93976

_ 90% CHD detection.
_ 90% DS detection.

Outcome analysis of HVRA's 'universal' fetal cardiac ultrasound program – 76811 with 2D fetal cardiac and cardiovascular color flow mapping 76825, 93325

10-year retrospective analysis of transfer rates of sick newborns with diagnosis codes of congenital heart disease delivered at Nyack Hospital demonstrated a transfer rate of 3-4 per 3,000 deliveries – significantly lower than the national average of 9-15 per 3,000 for similar-sized hospital (statistics obtained from Center for Disease Control.)

2008 prospective detection rate of cardiovascular disease

- 22 malformations were identified in the 2,718 cases studied for an incidence of 2.4 cases per 300 newborns.
- This compares to the most frequently quoted incidence of clinically significant congenital heart disease amongst newborns of 1:200 – 1:300.

II. AIUM's new cardiac outflow tract protocols – without fetal echocardiography – in the detection of fetal congenital heart disease, a literature review.

Interpretation of published studies must take into account the following factors that influence sensitivity (detection rate) of congenital heart disease.

Population studied – unselected versus low risk.

- Unselected population includes high-risk pregnancies including aneuploidy and/or extracardiac malformations.
- Low-risk population – attempts to exclude patients with traditional pretest risk factors i.e. family history, maternal disease, drug disclosure.

Gestational age at time of diagnosis – studies including CHD detection after 24 weeks exaggerate clinical value.

Single center versus multicenter studies – multicenter studies better reflect cross-sectional clinical practice.

There are no published prospective multicenter studies that establish the sensitivity of screening four-chamber and outflow tract protocols in the population in which congenital heart disease is most prevalent – in low-risk euploid pregnancies with no extracardiac malformations – the latter of which would increase cardiac scrutiny i.e. falsely increase screening sensitivity.

Background – the American published data prior to AIUM institution of outflow tract protocol.

Pinto. Barriers to prenatal detection of congenital heart disease: A population-based study. *Ultrasound Obstet Gynecol 2012*; 40:418-425.

A retrospective 10-year statewide (Utah) surveillance study of 1,474 cases of congenital heart disease between 1997-2007.

- Majority of CHD cases were missed prenatally and detection rates did not increase substantially during study period.
- 25% of CHD was missed amongst mothers who had one or more MFM ultrasound studies.

European literature review of CHD detection with four-chamber and outflow tract protocols but not fetal echocardiography.

Ogge. Prenatal screening for congenital heart disease with four-chamber and outflow tract views: A multicentered study. Ultrasound Obstet Gynecol 2006; 28: 779-784.

From Italy, in a multicentered prospective study of 4,873 low-risk patients

- 64% sensitivity for CHD (39 cases).
- Operators received specialized training.
- Gestational age at time of detection not stated.

Tegnander. Prenatal detection of heart defects in a nonselected population of 30,149 fetuses – detection rate and outcome. Ultrasound Obstet Gynecol 2006; 27: 252-265.

From Norway, in a prospective single center of greater than 30,000 unselected patients examined at 18-20 weeks –

- 44% sensitivity for isolated CHD
- 67% sensitivity for CHD in fetuses with abnormal karyotype and/or extracardiac malformations.

Most recent American publications on congenital heart disease detection with four chamber and outflow tract views – but not fetal echocardiography.

- D. Levey. Improved prenatal detection of congenital heart disease in an integrated healthcare system. *Pediatric Cardiology 2013*. 34:670-679.

  - 74% detection rate amongst 93 patients with significant congenital heart disease from a low-risk population yielding an incidence of 3.6 per 1,000 live births.

    - The live birth incidence of significant CHD quoted in this study is less than the 6 per 1,000 moderate-to-severe congenital heart disease cases expected based on an analysis of 62 studies published after 1955. J Am Coll Cardiol 2002 June 19; 39(12): 189-1900.

  - This article suggests the *possibility* of improvement in CHD detection employing outflow tract in comparison to the well-documented 30-50% detection rate in developed countries.

    - This study was performed within an idealized environment requiring dedicated training and multidisciplinary review of screening studies.

    - The 74% detection rate included fetuses with other extracardiac malformations and aneuploidy necessitating increased scrutiny of the heart.

  - Therefore even under idealized conditions a lower CHD detection rate could be expected for the majority of fetuses with CHD, i.e. as an isolated lesion in patients with no risk factors.

III. **AIUM's new outflow tract protocol as a *screening* tool does not necessitate the clinically most important interpretive *diagnostic* endpoint – reporting the exclusion of the five major patterns of congenital heart disease allowing safe delivery in a community hospital** – hypoplastic left heart, Tetralogy of Fallot, transposition of the great vessels, atrioventricular septal defect, aortic coarctation.

- AIUM's new outflow tract protocol remains an incomplete cardiovascular evaluation which ***does not*** include the following –
  - Evaluation of the cardiac valves for regurgitation and stenosis
  - Pulmonic veins for anomalous pulmonary venous return
  - Right subclavian artery – see below.

IV. **AIUM's updated 76811 outflow tract protocols remain an incomplete exam for genetic and syndromic evaluation.** Detailed obstetrical ultrasound without 2D cardiac 76825 and cardiovascular color flow mapping for genetic views has a 50-70% Down syndrome detection rate in comparison to a 90% detection rate when echocardiography is included.

The following vascular pathologies as aneuploidy and/or syndromic markers – are missed on AIUM's detailed 76811 protocol.

- Identification of an aberrant right subclavian artery and identification of normal course to the right subclavian artery has positive and negative Down syndrome likelihood ratios of 27 and 0.7 respectively
- Agenesis of ductus venosus
- Aberrant drainage patterns of ductus venosus and umbilical vein
- Mitral and tricuspid valve regurgitation

• Genetic sonography: the historical and clinical role of fetal echocardiography. Devore. *Ultrasound Obstetrics Gynecology. 2010*; 35: 509-521.
• Aberrant right subclavian artery: A marker for chromosomal abnormality. *Ultrasound Obstet and Gynecol. 2010*: 36:548-552. Borenstein.
• Aberrant right subclavian artery: incidence and correlation with other markers of Down syndrome in second trimester fetuses. *Ultrasound in Obstetrics & Gynecology. V39. Feb 2012*. Paladini. p. 191.
• The umbilical vein anomaly in fetuses with Down syndrome. *Ultrasound Obstetrics and Gynecol. 2010*: 297-301. Achiron
• Prenatal diagnosis of ductus venosus agenesis and its association with cytogenetics/congenital anomalies. *Prenatal Diagnosis*. 202: 995-1000.
• Scala. Aberrant right subclavian artery meta-analysis. *Ultrasound Obstet Gynecol. September 2015.*