Comment - Prof. Stuart Weiner

Jefferson University, Philadelphia, USA

I think that fetal echo, like all perinatal ultrasound, should continue to be two-tiered, with appropriate patient education and continuing advancement of the standards expected for both tiers. This is the way that all OB ultrasound has evolved over the past 60 years, and I do not feel that it should change. This will permit and stimulate continued evaluation of cost/benefits, better equipment, research, continuing education, and improved culturally- and financially-sensitive patient care.

Using fetal echo as a screening tool for DS sounds good, but only for those who can do it right, i.e., at the highest level. Since only 45-50% of DS fetuses have CHD and fetal echo will detect 90-95% of these, this will only identify 40% of the 1/500 fetuses with DS as being "high risk" for DS, perhaps after two fetal echo exams for all 500 of them. Then serum markers or more expensive cell-free DNA will follow, then expensive and more risky amniocentesis, expensive karyotype or more expensive microarray, then perhaps abortion for some of the 1/1000 fetuses identified to truly have DS. All of these cumulative expenses and risks (including procedure-related miscarriage for some euploid fetuses who might have benefited from pediatric cardiac surgery) must be considered and weighed against the economic and social costs of "missing" a DS diagnosis.

Perhaps we could explore augmenting a first-tiered fetal echo (at the standard general fetal anatomy scan by ISOUG standards) with the most cost-efficient pathway for further work-up and management of those fetuses with abnormal cardiac findings to improve the cost-benefit ratio of fetal echo as a screen for DS..

With the current DS screening advertised as 90% (Sequential Screen) or >99% (cffDNA) sensitive for DS, there is a very high expectation and consequent severe medico-legal implication if a DS is missed. The American health care system now places a supreme value on patient education and autonomy, but the plaintiff's attornies undermine that by flaunting the advertised sensitivity of screening for DS and always finding some flaw in the documented informed consent process.

In our MFM division, we have indeed missed CHD and DS, and we thoroughly review each such case in an objective, non-threatening, and educational fashion, always trying to improve, but never expecting or claiming perfection.

Comment - Maciej Słodki MD, PhD, Assoc. Professor

Department of Prenatal Cardiology, Polish Mother's Memorial Hospital Research Institute, Vice-president of the Association for Prenatal Cardiology Development, Secretary of Prenatal Cardiology and Fetal Echocardiography Section of Polish Ultrasound Society, Poland

The article by Respondek-Liberska entitled "Missing diagnosing of Down syndrome by prenatal ultrasound" is very interesting. I think that it is difficult to give simple answer for the questions in the form. Each case is individual and success depends on many aspects such as: how competent is the person performing an exam, what kind of exam was proposed to the patient and what kind of information did the doctor give to the patient, did he explain everything about his competence or did he inform the patient about another kind of exam that he doesn't perform. Did the exam report contain information about conditions of the exam, and finally conclusions and orders. Everything is very important and has a crucial meaning in potentially making a mistake.

Detecting a Down syndrome (DS) is still a challenge and as we can see there is a lot to do in this field of medicine but in my opinion we shouldn't concentrate our effort more on improving the detection of DS but on improving the detection of congenital heart disease (CHD). Congenital heart defects account for almost one per one hundred pregnancies and for one-third of all congenital anomalies and are the leading cause of infant mortality due to birth defects. CHD is eight times more likely to happen than DS. Moreover, almost half of DS cases has got CHD, so if we improve the detection of CHD we can automatically improve the detection of DS.

I agree with most of the questioned doctors that all sonographers performing obstetric scans should have a high degree of competence in detecting or suspecting the presence of a major fetal cardiac defect. Probably in the future it will be a group of people with perinatology specialization. Nowadays in Poland we have 508 doctors with certificate of competence on nuchal translucency (NT) scan (data from Fetal Medicine Foundation website: www.fetalmedcine.org) and only 52 doctors with competence of fetal heart examination, basic (n=45) and advance (n=7) (data from The National Registry of Fetal Cardiac Pathology, www.orpkp.pl). Referring to the article "Missing diagnosing of Down syndrome by prenatal ultrasound" by Respondek-Liberska we can see that in many cases detecting congenital heart disease can be a clue in detecting Down syndrome. The basic level should be based on sonographers performing obstetric scans and advanced level should be based on fetal cardiologist who consult patient and give proper diagnosis and prognosis which corresponds in 90% with postnatal diagnosis and prognosis. Tegnander and colleagues published in Ultrasound in Obstetrics and Gynecology their studies based on 30149 fetuses and showed that to obtain basic level experience in assessing four chamber view and 3 vessel view it takes about 5 years and gives a doctor a very good percentage of detecting CHD which amounts to 50 percent. Another study by Pezard and colleagues published in Prenatal Diagnosis proved that the most important thing which influences on ultrasonographers' training on prenatal diagnosis of CHD are not weekend or weekly courses but regular training in referral centers for diagnosing of CHD.

We have to realize that only 1 in 10 doctors with competence in NT scan has got the competence to perform basic fetal heart exam with 50% of detecting success.