MISSED DOWN SYNDROME AND CONGENITAL HEART DEFECT IN PRENATAL ULTRASOUND (US) - MALPRACTICE OR NOT? OPINIONS OF 12 EXPERTS

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Abstract

An example of missed Down syndrome with congenital heart defect by prenatal ultrasound evaluation was presented. A jury of 12 physicians, experts in prenatal ultrasonography and echocardiography were asked in questionare was this malpractice or not.

The answers were very different. The results of the questionaires were discussed with the background to the selected data from Eurocat, from Polish National Prenatal Cardiac Registry, from Polish Registry of Congenital Malformations by 2nd year of life, and financial data of the Polish Prenatal Program in Lodz Region.

Should we increase the cost of screening or the cost of ultrasound and echo training ? Or just provide patients with better knowledge regarding the differences between expertise of primary care obstetricians and experts in referral centers.

Key words: *Down syndrome, congenital heart defect, prenatal adn malpractice*

INTRODUCTION

The problem of a diagnostic mistake upon ultrasound examination is not new, however relatively rare discuss among the physicians in a scientific manner. Here is a an attempt to make such ana analysis.

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CASE AS THE SUBJECT OF ANALYSIS

A healthy 29-year old woman in her 2nd pregnancy, with a negative family history, and negative gynecological and medical history, remained under care of an obstetrician with fifteen years of clinical practice. The patient's first child was born healthy 2 years ago. Regular visits confirmed a normal course of pregnancy. Serum screening for fetal aneuploidy and NTD were negative. The ultrasound examinations were regularly performed and had revealed no abnormalities since the sixth week of pregnancy. At the time of the 11-14 week scan, the NT measured 1.8 mm, flow in the ductrus venosus was normal, and there was no tricuspid valve regurgitation. Collectively, she had 12 prenatal visits and 10 ultrasound examinations. The patient gave birth to a newborn which weighed 3500 g, APGAR 9 with typical phenotype of Down syndrome (DS) and a heart defect of complete atrio-ventricular septal defect. Mother and child were discharged home on the 5th day after delivery.

According to the classification of heart defects in prenatal cardiology, complete atrio-ventricular septal defect is a severe malformation but without the necessity of cardiac surgical intervention in the neonatal period. Cardiac surgery is performed most commonly at the 3rd – 6 th month of life, but DS remains.

METHODS AND MATERIALS

Twelve experts in prenatal ultrasound – physicians with a Basic Certificate of Fetal Heart Examination (of the Polish Ultrasound Society) or a Certificate of Echocardiographic Fetal Heart Examination (advanced) and laureates of Dr. Andrzej Respondek Prize - were asked to fulfill the questionnaire with 11 detailed questions.

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Questionaire

1. If the obstetrician had informed the pregnant woman that the likelihood of detecting a fetal cardiac malformation in his office was lower than 50%

A) he did not make a diagnostic error

B) he still made a mistake (missed Down syndrome)

C) it is hard to say

2. The obstetrician is obligated to perform a fetal heart examination. It is a separate diagnostic procedure performed by fetal cardiac refferal centers and pregnant women should know about this.

A) I do agree

B) I do not agree

C) it is hard to say

3. An obstetrician, after making such a severe mistake, should be retrained (at his/her own expense) during the next 6 months at the referral prenatal cardiology center type C (the highest referral center) to be able to discriminate normal from abnormal heart.

A) I do agree

B) I do not agree

C) it is hard to say

4. An obstetrician, following such a severe mistake (failure to detect ds or heart abnormality), should pay large damages to the family, or should cover social allowances for the handicapped child (answer A); or he should not cover any financial expenses, and those should be paid by his insurance (answer B)

A) I do agree

B) I do not agree

C) it is hard to say

5. An obstetrician has no obligation to detect congenital malformations. It is desirable if he/she can do so, but his major obligations is to take care of a pregnant woman and to deliver her baby in good condition. In this particular case, there was normal pregnancy and the fetus and neonate were in clinically stable condition.

A) I do agree

B) I do not agree

C) it is hard to say

6. Patients and pregnant women should be better informed regarding the spectrum of fetal echocardiographic examinations, and they should not demand detailed analysis of fetal heart from primary care obstetrician, as it is a separate procedure.

A) I do agree

B) I do not agree

C) it is hard to say

7. In my career in ultrasonography/echocardiography – "I have never made a mistake"

A) I do agree

B) I do not agree

C) it is hard to say

8. The detection of Down sydrome with a heart malformation is

A) very difficult

B) is relatively easy

C) I have no opinion

9. Ultrasonographic/echocardiographic errors should be unveiled, discussed and analyzed in order to learn from them rather than repeat them.

A) I do agree

B) I do not agree

C) it is hard to say

10. Ultrasonographic/echocardiographic errors are made so rarely, that since we perform hundreds of examinations, we should not discuss them so extensively. They are inherent to medical practice for all of us, and they are the personal problem of every physician.

A) I do agree

B) I do not agree

C) it is hard to say

11. Did the obstetrician under discussion:

A) make an error and he should therefore suffer the consequences?

B) did not make an error and he should not suffer the consequences?

C) I have no opinion.

RESULTS

Results of the analysis of the questionnaire (with initials of jury members) are given in Table 1.

Question 1 (Q1). The majority (7/12; 60%) choose answer A, which stated that if the obstetrician had informed the pregnant woman that the likelihood of detecting a fetal cardiac malformation by his examination was lower than 50% and fetal heart examination is a separate examination performed by an expert, he did not make a diagnostic error.

Q2. The majority (8/11) choose answer B, suggesting that an obstetrician should perform a fetal heart examination.

Q3. The majority choose answer A, which stated that an obstetrician after making such a severe mistake should be re-trained (at his/her own expense) during next 6 months at the centre of prenatal cardiology type C (the highest referral center) to be able to discriminate a normal from abnormal heart.

Q4. The majority answered „B": to the following statement an obstetrician following such a severe mistake (failure to detect ds or heart abnormality) should pay large damages to a family, or should cover social allowances for the handicapped child (answer A); or he/she should not cover any financial expenses while those should be paid by his insurance (answer B).

Q5. To the statement „an obstetrician has no obligation to detect congenital malformations. It is desirable if he/she can do so but his major obligation is to take care for the pregnant woman; in case under discussion, both fetus and newborn were fully cardiologically viable" . The majority answered that they disagree with it.

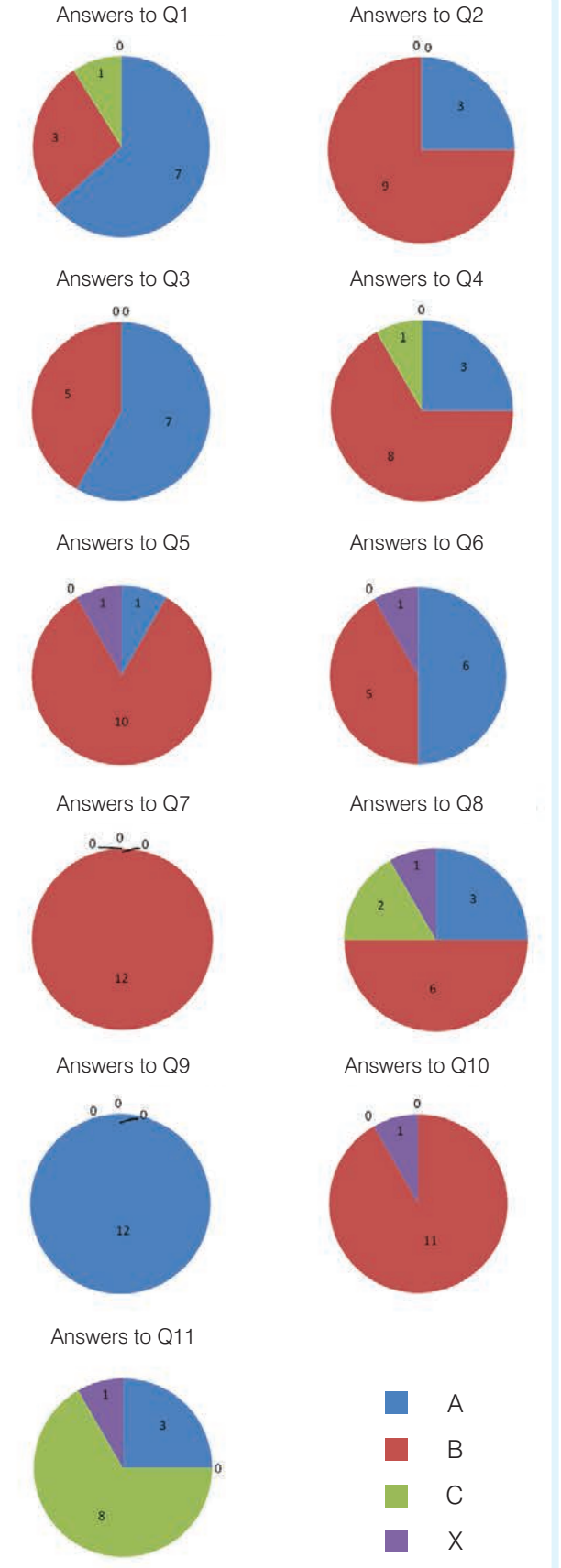

Chart 1. Graphical presentation of answers (A, B, C or X meaning "I have a different opinion") for questions (A & Q)

Q6. To the statement "Should pregnant women be better informed about the spectrum of fetal cardiac examination available for their fetus? However, they should not demand a detailed analysis of the fetal heart as it is a separate procedure", a slim majority answered „I agree" (6 x A, but 5 x B).

Q7. The question concerned making a mistake in his/her career in ultrasonography/echocardiography – "I have never made a mistake"; all answered that this is not true.

Q8. The question concerned the difficulty of detecting DS and a heart malformation – A: such a detection is very difficult; B: is relatively easy; C: I have no opinion, I have never met such a case during my career". Only 3 persons choose „A" and 6 persons choose „B".

Q9. The question concerned ultrasonographic/echocardiographic errors and whether they should be unveiled, discussed and analyzed to learn from them and not repeat them. 100% answered that they agreed with the statement.

Q10. The question concerned the statement „ultrasonographic/echocardiographic errors are made so rarely, while we perform hundreds of examinations, that we should not discuss them so extensively. They are inbuilt into medical practice of all of us and they are the personal problem of every physician". Eleven answered they do not agree with this statement.

Q11. Did the obstetrician under discussion: make an error and should suffer the consequences (answer A); did not make an error and should not suffer the consequences (B); I have no opinion (answer C). The majority (8/11) choose answer C.

DISCUSSION

Current data on the prevalence of DS in Europe stem from the Eurocat program and covered 7044 cases of DS in 28 European registries between 2000-2010. This data suggests, that despite introduction of screening for DS in many countries, the prevalence does not change. Moreover, they confirmed that in 43% of newborns with DS, congenital heart defects are detected and in another 15%, extracardiac malformations are found[1].

Data from Poland[2], confirm the above mentioned observations and stress that cardiac malformations occur in almost every second case of DS.

If the financial aspect of prenatal diagnosis of DS is taken into consideration within the framework of the Program of Prenatal Diagnosis, according to the data published by the Lodz branch of National Health Fund (NFZ, in Polish), the cost to diagnose DS (and the other most frequent genetic syndromes) between 2011 and 2013 was very high and revealed a raising trend from 54 486 Polish Zloty (approximately USD 16,000) to 67 540 PLN (USD 20,000) per one case (Table 3). If only a proportion of those costs of the Program of Prenatal Diagnosis was allocated to improve prenatal cardiologic diagnosis, it might be possible to obtain more reliable results at lesser cost . Even if pregnant women would be offered

echocardiographic examination twice during a single pregnancy, those costs would not exceed an additional 1000 PLN (USD 294) per patient[3].

However, Polish residents in obstetrics and gynecology do not learn prenatal cardiology. As a result, not only grave errors are made; but those errors are met with a substantial tolerance from obstetricians. We already discussed diagnostic errors of non-detection or late detection of DS in our journal[4] but everyday practice shows it is still a hot topic. Sonographic markers of DS have been well described in the past[5-7,] and they became standard textbook entries[8,9].

In the Pubmed database there are 1542 entries on the prenatal diagnosis of DS but only 6 on DS and the prenatal diagnosis of cardiac malformations, including one from our centre[3]. This may suggest that prenatal cardiology for screening for DS is still underestimated. Theoretical knowledge is different from practical applications[10]. The analysis under discussion suggests that for such an apparently simple problem, if an obstetrician made a diagnostic error with consequences that influence the future life of the patient, there is not a consensus opinion.

Q & A 1. Althogh the majority did agreed with my suggestion, the answers were not fully satisfactory, because.... not all surveyed experts thought that written information presented to pregnant women was mandatory and such information is not only indicated but the mandatory may reflect a paper from "Law and Medicine" section of Prenatal Cardiology; September 2014 issue.[x] A patient sued her obstetrician because he did not detect fetal heart malformation of Double Outlet of the Right Ventricle. However, an obstetrician recorded in a patient's files a „normal 4-chamber view", which could be a proper diagnosis at the 20th week of gestation, but he also documented that he had informed the pregnant woman about the availability of a more specialized echocardiographic examination which may increase chances of detecting a heart malformation. Such a written note in the patients' file was his insurance policy.

Q & A 2. The majority of answers were compatible with the statement that an obstetrician should evaluate a fetal heart. At the congress of International Ultrasound in Obstetrics & Gynecology Society in Zagreb 3 indications for fetal heart evaluation had been listed: 1) pregnancy; 2) pregnancy and 3) pregnancy. However, we still talk too little and write too little on the differences between fetal heart evaluation performed by an obstetrician and by prenatal cardiologist with appropriate training and certification. From the viewpoint of diagnostic organization and the National Health Fund in Poland which paid for it, in the signed contract, only a single level of payment is itemized, irrespective of the competence level. It is common knowledge how much time and effort is spent to confirm normal fetal heart structure not to mention to detect a fetal heart malformation, to perform all the measurements, to establish fetal circulatory function, to consult the family and finally, compose heart diagram.

Q & A3. The majority (58%) of surveyed experts agreed that in the case of making a diagnostic error in the field of prenatal cardiology, an obstetrician under discussion should refresh his/her knowledge with appropriate further education. However, 42%

	Physician	1	2	3	4	5	6	7	8	9	10	11
1	MRL	A	A	A	A	A	A	B	A	A	B	A
2	LD	A	B	B	C	B	A	B	A	A	B	C
3	MS	A	B	A	B	B	A	B	B	A	B	C
4	P D	A	A	B	A	B	A	B	A	A	B	A
5	PA	B	B	A	B	B	B	B	B	A	B	A
6	KL	B	B	A	B	B	B	B	B	A	B	A
7	PG	A	A	B	B	B	A	B	C	A	B	C
8	JP	A	B	A	B	B	A	B	B	A	B	C
9	AD	x	B	B	B	x	x	B	B	A	B	C
10	PS	B	B	A	B	B	B	B	C	A	B	C
11	PK	C	B	A	A	B	B	B	B	A	B	C
12	MK	A	B	B	B	B	B	B	x	A	x	x
		7A	9B	7A	8 B	10 B	6A	12 B	6B	12A	11 B	7 C
		3 B	3 A	5 B	3 A	1 A	5 B		3 A			4 A
		1C			1C				2C			
		1 X				1 X	1 X		1 X		1 X	1 X

Table 1: Answers for questionaire (answers: A, B, C and X - meaning "I have a different opinion")

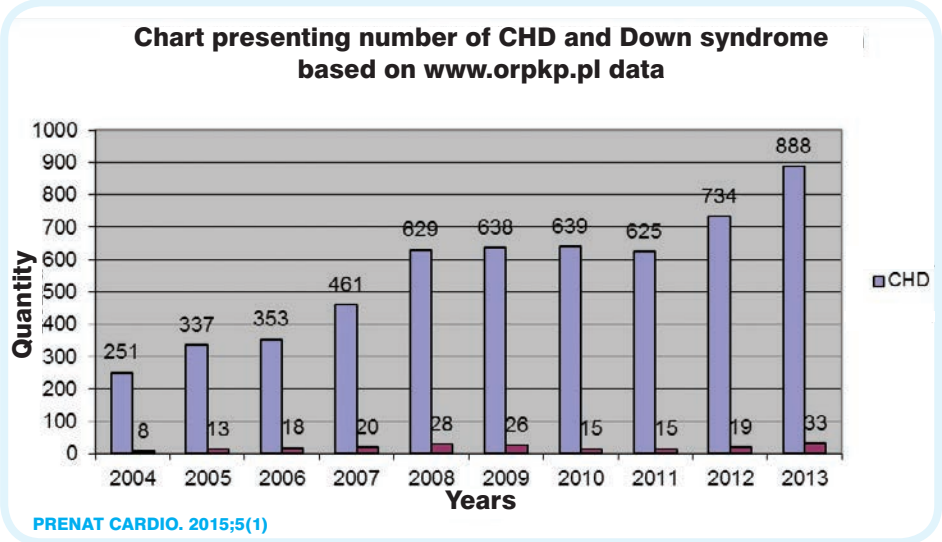

Table 2: Data from Polish National Registry of Prenatal Cardiac Anomalies from years 2004-2013 (red bars: congental heart defect and Down syndrome), blue bars: numer of fetuses with heart defects

Years	2004	2005	2006	2007	2008	2009	2010	2004-2010
Number of newborns with DS. (live born and in utero demises)	453	403	337	458	454	396	357	2858
Number of newborns with DS. and congenital heart defect (live born and in utero demises)	158	147	122	156	171	170	163	1087

Table 3 . Number of newborns with Down syndrome born alive and dead in Polish Registry of Congenital Malformations in 13 voivodoships in Poland (dolnośląskie, kujawsko-pomorskie, lubelskie, lubuskie, łódzkie, mazowieckie, opolskie, podkarpackie, pomorskie, śląskie, warmińsko-mazurskie, wielkopolskie i zachodniopomorskie) in years 2004-2010 and 3 others (małopolskie, podlaskie i świętokrzyskie) in years 2007-2010 (data from prof. A. Latos-Bieleńska)

of surveyed experts thought otherwise. Those experts did not specify whether they were against additional training or against the suggestion that costs of such a training should be met by the obstetrician.

Q & A4. 67% of evaluated experts were against meeting any consequences of erroneous diagnosis. I, personally, would prefer to be insured and to have a feeling of „legal security" in case of patient's accusation than to pretend that nothing happened. If I occupy a real position of trust from in the viewpoint of a patient, and if I spent my time and expertise, but I made a mistake, I must expect consequences. In the Polish movie Gods (2014), the main character, a famous cardiac surgeon, experienced the death of his patient, a girl with a complex heart malformation. He defended himself in front of the Ethics Committee, and was acquitted, but he suffered a psychological toll from this unsuccessful surgery for a long time and only „absolution" from the girl's mother allowed him to regain composure. „Perhaps this was the will of God", said the girl's mother, but not all patients think the same, and we all know that medical liability claims have a tendency to be higher every year.

Q & A5. Most agreed with the statement that an obstetrician/ultrasonographer should be able to detect fetal malformations and not only care for a pregnant woman in general terms. But my professional life verified this statement. It is relatively easy to detect cardiac or extracardiac malformation at the 3rd trimester, in particular at the reference center. But to detect the same malformations at the 1 st or 2nd trimester, especially at a low risk „screening" office, when a pregnant woman may still legally ask for termination of pregnancy, is much more difficult. It was published about the possibility of a changing phenotype between the 2nd and 3rd trimester in our journal.[11] It must be stressed that signs of DS may also change throughout fetal development.[12]

The majority of pregnant women in Poland, despite an unfavorable diagnosis, decide to continue the pregnancy (www.orpkp.pl), but modern medicine and Polish law do not demand such a stoic attitude. Not all women must or are able to withstand the burden of giving birth to a physically or mentally malformed child. Polish law, similar to the majority of European laws, offers a choice. However, this choice is not always supported by the possibility to make a proper diagnosis in time.

Because of this, i personally support a new form of health care organization for pregnant women, in which the ultrasound examination is performed by an obstetrician who is well prepared to care for a pregnant woman, to follow the influence of pregnancy on her clinical status, who is able to evaluate biometrics and fetal sex and in the majority of cases, blood flow in the umbilical and middle cerebral arteries. And for those pregnant women, who do not consider termination of pregnancy because of any abnormalities, such care seems totally sufficient.

But those pregnant women who want to give birth only to a healthy baby and those who would consider termination of pregnancy in the case of detection of a severe fetal malformation, should be referred to centers that specialize in fetal diagnosis and therapy and which provide above average expertise. In Poland, there are more and more such centers under the name of Outpatient clinics or Departments of Diagnosis and Therapy of Fetal Malformations.

Q & A6. Fifty percent of surveyed experts (50%) agreed that pregnant woman should be better informed on the spectrum of prenatal diagnoses, including prenatal cardiology. However, the experts panel thought that pregnant woman are informed sufficiently. I was asked many times by pregnant patients why they did not receive any information regarding the fetal heart from their obstetrician but were informed by "Dr. Google" instead. It is doubtful that the patient herself discriminated between basic fetal heart evaluation and full fetal heart examination; even the Polish National Health Fund does not.

Q & A7. This question was, obviously tricky, as it is common knowledge that prenatal sonography in general, and

		Year		
		2011	2012	2013
Number of women		3634	3982	4303
Number of invasive procedures	amniopunction	394	448	
	CVS	44	45	
Malformations seen in US		60	47	43
Malformations after invasive procedures		60	49	45
Total costs from the National Prenatal Programme		3 269 199,46 PLN	3 229 440,96 PLN	3 039 333,64 PLN
Preis for detection of 1 case		54,86 PLN	65,906 PLN	67,540 PLN

Table 4: Data about the costs of Down syndrome detection in Lodz Region based on Regional National Health System data

echocardiography in particular are liability minefields, with a significant risk of problems or omissions. We even know the reasons but this Q7 was a preface to Q8. Answering Q8, merely 2 surveyed experts (including myself) answer that to detect DS prenatally may prove difficult even when common atrio-ventricular canal accompanies DS. On the other hand, it is relatively common to make a false positive diagnosis of DS because of an enlarged NT, abnormal triple test, shortening of long bones, enlarged renal pelvis, two-vessel umbilical cord and… a healthy newborn is born. It is well known that the presence of the so called soft markers are not fully diagnostic of DS, as each has a low sensitivity[13]. In contrast, it is easy to diagnose prenatal trisomy 13 or 18 [15] with ultrasound alone wherein, since the 1st trimester, multiple anomalies are seen as well as abnormal fetal growth. But in case of DS, dysmorphic features may not be obvious in the 2D, 3D or 4D prenatal examination; the fetus may have normal structure of the heart or cardiac malformation in the form of ASD ostium primum. The latter may be practically unobtrusive in the 1st or second trimester. It is not unusuall to detect common atrio-ventricular canal in the 3rd trimester by expert erroneously missed in the 1st or 2nd trimesters by obstetrician.

Q & A9 and ***Q & A10.*** All surveyed experts agreed that sonographic errors are important caveats of everyday work.

In the Department of Prenatal Cardiology in our institution, such errors are rarely made. And if we do make an error, we spend a lot of time discussing them at our meetings, knowing that retrospective analysis is part of our education

However, despite this apparently easy to solve puzzle which comes down to the basic question "is the obstetrician under discussion guilty or not guilty of failure to detect a fetus with DS", the majority of experts evaded the answer and choose "I have no opinion".

As a result, we conclude that this remains a very difficult problem and such a questionnaire did not solve it. In my mind, I must stress it again and again, prenatal diagnosis of DS is very difficult but feasible at the reference centre which deals with fetal malformations (at least 100-150 cases a year). The accurate results are obtained in more than 90% of cases but still it is not 100%. In contrast, for the general obstetrician/ultrasonographer, who sees only 2 – 3 malformations yearly, detection of DS may be extremely difficult.

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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.