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EVALUATION OF FETAL ANOMALIES BY TWO- AND THREE-DIMENSIONAL ULTRASOUND

PROSUDBA FETALNIH MANA RAZVOJA DVO- I TRO-DIMENZIONALNIM ULTRAZVUKOM

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Original paper

Key words: fetal anomalies, prenatal diagnosis, three-dimensional ultrasound

SUMMARY. ***Objective.*** To assess clinical impact and the potential of three-dimensional ultrasound (3D US) for prenatal evaluation of fetal malformations compared to two-dimensional ultrasound (2D US). Clinical impact of 3D US in the cases with anomalies were determined. ***Methods.*** Seventy-six patients with 190 anomalies were scanned prospectively with both 2D and 3D US by two fetal medicine specialists who have extensive experience in 3D US. Each anomaly was reviewed again to determine whether the 3D US data were advantageous when compared to 2D US images. ***Results.*** The 3D US images provided additional information in 130 (69%) anomalies, were equivalent to 2D US images in 60 (31%) anomalies and were not disadvantageous in any of the cases. The 3D US was particularly helpful in evaluating fetuses with craniofacial anomalies, spinal defects, thoracic abnormalities, body surface and extremities. ***Conclusion.*** In comparison with 2D US, 3D US offers more diagnostic information in evaluating fetal malformations, particularly in displaying fetal malformations of the face and cranium, spine, extremities, and body surface. 3D US is an excellent adjunctive tool to 2D US in the evaluation of fetal anomalies.

Izvorni članak

Ključne riječi: fetalne anomalije, prenatalna dijagnostika, trodimenzionalni ultrazvuk

SAŽETAK. ***Cilj.*** U usporedbi s dvodimenzionalnim ultrazvukom (2D UZ) procijeniti klinički značaj i mogućnost trodimenzionalnog ultrazvuka (3D UZ) za prenatalnu prosudbu fetalnih mana razvoja. Odrediti klinički značaj 3D UZ u trudnoćama s anomalijom. ***Metode.*** Dva specijalista iz fetalne medicine, s posebnim iskustvom u 3D ultrazvuku, prospektivno su pomoću 2D i 3D ultrazvuka pregledavali 76 trudnica/fetusa sa 190 anomalija. Svaka je anomalija bila ponovno vrednovana glede prednosti 3D ultrazvuka u usporedbi s 2D ultrazvukom. ***Rezultati.*** 3D UZ slike su pružile dodatnu informaciju kod 130 (69%) anomalija, bile su podjednake valjanosti u 60 (31%), a nisu bile manje valjane ni u jednom slučaju. 3D UZ je bio posebice pomoć u otkrivanju plodova s anomalijama lica i glave, defektima kralježnice, abnormalnostima prsišta, površine tijela i ekstremiteta. ***Zaključak.*** U usporedbi s 2D ultrazvukom, 3D ultrazvuk pruža veće mogućnosti za otkrivanje fetalnih malformacija, posebice u prikazu anomalija lica i glave, kralježnice, ekstremiteta i površinskih promjena, 3D UZ je u otkrivanju fetalnih anomalija izvrsna pomoć dvodimenzionalnom ultrazvuku.

Introduction

It is clear that one of the great advantages of 3D ultrasound is that the information remains captured as a volume and it is possible to reconstruct the recorded image and modify all the adjustments as if the patient was still present.[1,2] This enables us to manipulate the image, re-rotate it three-dimensionally and achieve another 3D reconstruction from the data already taken.[3] There are also electronic scalpels, which assist by cutting off and eliminating all parts that can hide or distort the area we wish to study. With these facilities, we can improve the 3D reconstruction of an image that is less favorable by 2D US.[3,4] 3D US can observe a region of interest from any orientation and it obtains a volume data set composed of a series of 2D images and thus is able to conveniently demonstrate the features of the lesions and their spatial relationships. The different display modalities like transparency mode, maximum intensity mode, surface rendering mode enable operator to evaluate the pathology in the structures of the fetal cranium/face, spine, extremities, and body surface. Multiplanar mode can be used to observe internal structures from various orientations simultaneously.[4,5]

The purpose of this study was to compare 2D US and 3D US in the evaluation of fetal malformations. Clinical impact of 3D US and pathologic and clinical outcome of the cases were determined.

Material and methods

This study was performed prospectively from January 2005 to June 2006. One thousand twenty-four pregnant women with high-risk pregnancies or with anomaly suspicion were referred to the Fetal medicine unit of Marmara University Hospital. Among these women, 76 patients who were confirmed to have abnormal fetuses with a total of 190 abnormalities were included in the study. All patients with an anomaly diagnosis were underwent 3D and 4D US evaluation of the fetus after 30–45 minutes 2D US evaluation by switching the machine to 3D mode. All of the scans were performed and reviewed by the same two extensively experienced fetal medicine specialist on 3D US. Each anomaly was re-

Table 1. Comparison of 2D US and 3D US findings in 190 malformations in 76 fetuses with postnatally confirmed abnormalities
Tablica 1. Usporedba 2D i 3D ultrazvučnih nalaza 190 mana razvoja u 76 plodova s postnatalno potvrđenom abnormalnošću

Body region – Abnormality *Područje* – Abnormalnost	No. Broj	Advantageous Prednost	Equal Podjednako
CNS			
Holoprosencephaly	8	6	2
Hydrocephalus	10	2	8
Unilateral ventriculomegaly	3	2	1
Mild ventriculomegaly	8	6	2
Encephalocele	3	3	0
NTD, Spina Bifida	10	10	0
Meningomyelocele	4	4	0
Dandy-Walker	4	3	1
Microcephaly	1	0	1
Choroid Plexus cyst	6	4	2
Galen vein aneurysm	1	1	0
Inferior vermian agenesis	3	2	1
	61	43	18
		70,49%	29,51%
Face – Lice			
Cleft lip/palate	2	2	0
Hypotelorism	2	1	1
Flat face	2	2	0
Single nostril	1	1	0
Absent nasal bone	2	2	0
Facial dysmorphism	1	1	0
Low set ears	2	2	0
Micrognathia	1	1	0
	13	12	1
		92,31%	7,69%
Neck – Vrat			
Fetal goiter	1	1	0
Cystic hygroma	4	3	1
Increased nuchal edema	2	0	2
Hydrops fetalis	3	3	0
	10	7	3
		70,00%	30,00%
Skeleton			
Thanatophoric dysplasia	1	1	0
Majewski Syndrome	1	1	0
Scoliosis	1	1	0
Short limbs	3	2	1
Clubfoot	4	4	0
Syndactyly	1	1	0
Clenched hands	1	1	0
Sandal Gap	1	1	0
Polydactyly	2	2	0
	15	14	1
		93.3%	6.7%
Thorax			
CCAM	2	2	0
Pleural effusion	4	4	0
	6	6	0
		100,00%	0,00%

Body region – Abnormality *Područje* – Abnormalnost	No. Broj	Advantageous Prednost	Equal Podjednako
Heart – Srce			
Fallot tetralogy	1	1	0
Hypoplastic left heart	3	2	1
Transposition	1	1	0
Ebstein anomaly	1	1	0
VSD	3	2	1
AVSD	2	2	0
Hyperechogenic focus	5	3	2
Double outlet right ventricle	1	1	0
Pericardial effusion	3	2	1
Cardiomegaly	4	0	4
	24	15	9
		62,50%	37,50%
Abdomen			
Omphalocele	3	3	0
Gastroschisis	3	3	0
Diaphragmatic hernia	1	1	0
Duodenal atresia	2	2	0
Hyperechogenic bowel	4	0	4
	13	9	4
		69,23%	30,77%
Genito-urinary			
Hydronephrosis	10	2	8
Duplex kidney	2	2	0
Hydroureter	2	1	1
Single kidney	1	1	0
Horseshoe kidney	1	1	0
Megacystis	1	1	0
Infantil polycystic dysplastic kidney	1	0	1
Renal agenesis bilateral	1	0	1
Posterior urethral valves	3	2	1
Renal cyst	2	2	0
Multicystic dysplastic kidney disease	1	0	1
Ectopic pelvic kidney	1	1	0
Hyperechogenic kidney	1	0	1
Ambiguous genitalia	2	2	0
Hydrocele	2	2	0
	31	17	14
		54,84%	45,16%
Miscellanous – Razno			
Nonimmune hydrops	2	2	0
Umbilical cord cyst	2	0	2
Single umbilical artery	10	2	8
Conjoined twins	1	1	0
Lumbar hemangioma	1	1	0
TTTS	1	1	0
	17	7	10
		41,18%	58,82% 1

VSD: Ventricular Septal Defect; AVSD: Atrioventricular Septal Defect; TTTS: Twin to twin transfusion syndrome.

viewed again to determine whether the 3D US data were advantageous when compared with 2D US images. Consensus was obtained for the diagnosis of each malformation. The evaluation of the fetal abnormalities both by 2D and 3D US is a routine in our clinic. All the patients were given informed consent to participate the study and they all were extremely pleased to be evaluated by 3D US as well.

All 2D and 3D examinations were performed by using Voluson 730 Expert Diamond II (GE Medical Systems, Milwaukee, WI, USA) equipped with a transabdominal 5 MHz and transvaginal 5–9 MHz 3D transducer. The volume data and the rendered 3D images were stored in digital form on an external hard disk (250 GB USB 2.0 hard drive; LaCie, USA) for rendering and re-analyzing. Information about the clinical outcome of the patients was obtained through the hospital records and direct phone calls to the parents. Malformations were confirmed by postnatal or postmortem follow-up. The results were evaluated by percentage calculation.

Results

The mean age of the women was 28 years (range, 17–42 years), and the mean menstrual age of the fetuses was 31 weeks (range, 11–38 weeks). Five women had twin pregnancy.

Seventy-six patients had a total of 190 anomalities that were evaluated by both 2D and 3D US. While 3D US images provided additional information and confirmed diagnoses in 130 (69%) anomalies, 2D US images equivalently confirmed the diagnosis in 60 (31%) anomalies and were not disadvantageous in any of the cases. The parallel validity i.e. the advantage of 3D US diagnostics for everyone of the 190 diagnosed fetal anomalies is presented on the *Table 1.*

Discussion

Although conventional 2-dimensional sonography (2D US) is able to detect many kinds of fetal malformations, some studies[6] have shown that the 2D US detection rate for fetal anomalies in low-risk pregnancies is poor in the primary care setting. Even in tertiary-care centers, only 40–50% of cases of fetal malformations are detected prenatally by 2D US.[6,8] Therefore, the adjunctive use of 3D US will increase the detection rate and quality of assessment of fetal anomalies.[8] Moreover the demonstration of a fetal defect in the third dimension is impossible by the conventional technique. Three-dimensional ultrasound allows visualization of the fetal malformations in all three dimensions at the same time, providing an improved overview and a more clearly defined demonstration of adjusted anatomical planes. In many studies it has been shown that 3D US was able to discover some complicated fetal malformations that were missed by 2D US and to determine the fetal malformations more precisely than 2D US did.[8,9] The advance of 3D volume sonography is enormously exciting and gives us control over the subject we are imaging, which goes well further than 2D imaging.[10] We can manipulate the volume, rescan the fetus using the volume, and display any part of it. However, we have much to learn to determine where the applications will be as this technology leaps forward in the future.[11] The 3D technique is superior to the traditional two-dimensional biometries, without the common limitations such as fetal position.[12]

Figure 1. 3D surface rendered US image of occipital encephalocele at 33 weeks *(left)*. Postmortem picture lateral view *(right)*. This image is presented to demonstrate the ease with which the 3D display enables pinpointing of the location of the bony gap in the skull through which brain protrudes

Slika 1. 3D UZ slika okcipitalne encefalokele s 33 tjedana *(lijevo)*. Postmortem postranična slika *(desno)*. Slika je prikazana da pokaže lakoću kojom 3D omogućuje prikaz lokacije koštane pukotine lubanje kroz koju se mozak izbočuje

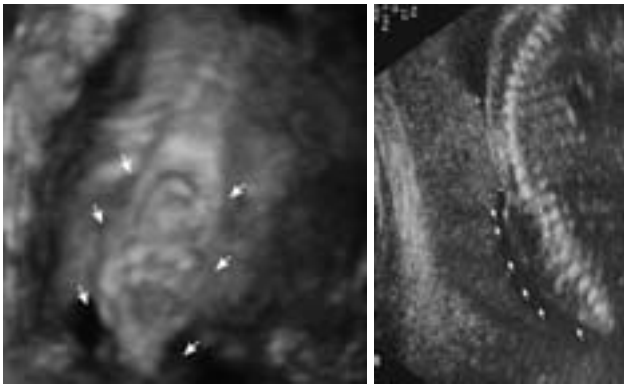

Figure 2. 3D surface rendered image of the open spina bifida in a fetus at 24 weeks of gestation *(left)*. 2D US image sagital section *(right)* showing spinal defect at 23 weeks' gestation

Slika 2. 3D površinska slika otvorene spine bifide fetusa s 24 tjedana trudnoće *(lijevo)*. 2D UZ sagitalna slika koja pokazuje manjak na kralježnici s 23 tjedna trudnoće *(desno)*

Merz et al.[13] examined 204 patients with 3D US and proved that 3D US is advantageous in demonstrating fetal defects. Hui-Xiong et al.[8] assessed the fetuses with abnormalities both by 2-dimensional (2D) and 3-dimensional (3D) sonography and has shown that 3D US definitely diagnosed all the abnormalities in 38 fetuses (93%), whereas 2D US did so in only 32 fetuses (78%). In 35 (60%) of the 58 malformations revealed by both 3D US and 2D US, the former provided more diagnostic information than the latter. Baba et al[12] reported that 3D US could demonstrate clearly malformations of the fetal face, ears, and fingers, which may be hard to visualize on 2D US. Dyson et al.[9] scanned 63 patients with 103 anomalies and thought that 3D US offered diagnostic advantages and had effect on patient management in 5% of cases. All these findings are similar to our findings. 3D US improves the diagnostic capability by offering more diagnostic information than 2D US in evaluating fetal malformations, particularly in displaying fetal malformations of the cranium and face, spine and extremi-

Figure 3. Three-dimensional multiplanar surface rendered image of the fetal face and neck at 28 weeks demonstrating fetal goiter very clearly *(left)*. Neonate with congenital goiter *(right)*

Slika 3. 3D multiplanarna površinska slika fetalnog lica i vrata s 28 tjedana trudnoće, koja vrlo jasno prikazuje fetalnu gušu *(lijevo)*. Novorođenče s gušom *(desno)*

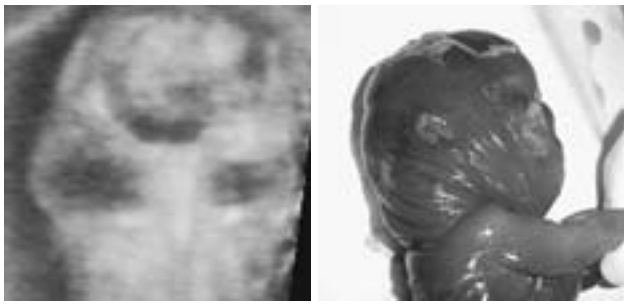

Figure 4. Bilateral cystic hygromata with diffuse edema in a fetus at 15 weeks of gestation by 3D scan *(left)*. The fetus after abortion *(right)*. Karyotyping revealed this baby as Turner syndrome

Slika 4. Obostrani cistični higromi s difuznim edemom u fetusa s 15 tjedana trudnoće prikazani 3D ultrazvukom *(lijevo)*. Plod nakon pobačaja *(desno)*. Kariotipizacija je potvrdila Turnerov sindrom

ties, and body surface.[8,9,13] Three-dimensional sonography is a valuable adjunct to 2D US in prenatal diagnosis.[13]

In the present study 3D US images provided additional information in 43 of 61 cases (70,49%) of CNS anomalies including holoprosencephaly, hydrocephalus, encephalocele *(Figure 1),* neural tube defects. In the cases of spina bifida we localized the spinal defects better by 3D US than 2D US scanning by using simultaneous multiplanar imaging and referencing to the volume rendered image in all of the 10 cases with spinal defect *(Figure 2)*. The 3D US offered advantages over 2D US imaging in terms of data storage as well. Volume data were stored on removable hard disks allowing for review with postprocessing capabilities and manipulation of the images by multiple physicians after the patient left the clinic.

Three-dimensional ultrasound offers the possibility of studying the fetal face and neck in a more global way than conventional two-dimensional ultrasound *(Figure 3,4)*. This part of the fetal body was and still is the most documented structure in 3D ultrasound.[14,15] A diagnosis of a cleft of the palate and lip are at times hard to make using 2D ultrasound. Using multiplanar mode, it is possible to evaluate orthogonal planes and axial planes simultaneously. Therefore the alveolar ridge and upper lip can be evaluated at the same time.[16] Likewise in our study, 3D US images were advantageous in 12 of 13 anomalies in detecting abnormal fetal profile and facial abnormalities mostly by surface rendered images. Three-Dimensional US clearly delineated the range and location of the cleft lip, and in the transparent mode, 3D US suggested the cleft palate. Lee et al.[14,17] analyzed 7 fetuses with confirmed facial cleft anomalies and found that surface rendering mode of the face may allow increased diagnostic confidence for normal and abnormal lips. Cephaloceles have been located and described better than by 2D ultrasound.[18] The cranial sutures and fontanelles were also successfully evaluated using 3D ultrasound.[19] Monteagudo et al.[20] worked on 24 fetuses with brain pathology.

Three-Dimensional ultrasound technology can effectively be used to examine the fetal brain. The ability to simultaneously view and review a brain volume in all three scanning planes, by navigating back and forth through digitally stored data was found to be clinically important.

Evaluation of the abnormality by 3D US in fetal skeleton and extremities using X ray mode and the maximum mode the bony structures such as bones and extremities can be visualized.[21] Skeletal dysplasias also can be evaluated by 3D US in conjunction with 2D US success-

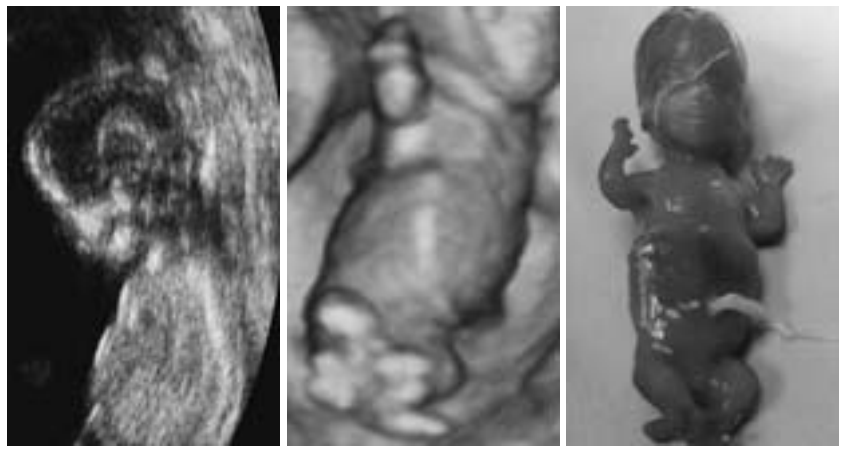

Figure 5. Thanatophoric dysplasia at 16 weeks. Sagital scan by 2D US shows narrow chest *(left)*, 3D surface rendered image of the fetus *(middle):* note marked difference between chest and abdomen; all the extremities are clearly seen to be extremely small by 3D US. The baby after abortion *(right)*

Slika 5. Tanatoforična displazija sa 16 tjedana trudnoće. Sagitalni presjek 2D ultrazvukom prikazuje usko prsište *(lijevo)*. 3D površinska slika fetusa *(sredina)*: izrazita razlika između prsišta i trbuha; svi su ekstremiteti uočljivo mali. Plod nakon pobačaja *(desno)*

Figure 6. 3D surface rendered image of a normal male genitalia *(left)* and multiplanar view of the hydrocele in a fetus at 39 weeks *(right)*. Abnormalities of the fetal gender are also easy to detect by 3D US

Slika 6. 3D površinska slika normalnog muškog spolovila *(lijevo)*. Multiplanarni izgled hidrocele u fetusa od 39 tjedana *(desno)*. Abnormalnosti fetalnog spola je također lako otkriti 3D ultrazvukom

fully.[22] Developmental anomalies of the skeleton are identified with a high degree of reliability.[23,24] The vertebrae, ribs, and clavicle can be demonstrated as well as the general appearance of the fetal body.[22–25] As far as the extremities are concerned, 3D ultrasound is almost the ideal tool to evaluate the hands and feet. It performed better than 2D ultrasound in evaluating fetal digits. It can accurately delineate flexion deformities such as clubfoot and overlapping fingers.[26,27] In agreement with the literature skeletal anomalies were better visualized by by 3D than 2D scanning in 14 of 15 anomalies in our study *(Figure 5)*. Especially transparent maximum mode and surface rendered images of the fetus by rotating the rendered volume images clearly documented the anomalies.

Anterior abdominal wall defects (omphalocele, gastroschisis), congenital diaphragmatic hernias, intraabdominal and intrathoracic masses, their composition, vascularity, inner wall of the cysts can be advantageously evaluated with 3D surface rendered and multiplanar modes.[4,28–30] However some authors suggested that 3D US added little value in diagnosing malformations of the fetal thorax and abdomen.[1,8] Among 6 thoracic anomalies evaluated 3D US were advantageous in all of them. But this was probably due to our small anomaly number on this part of the fetus.

Fetal sex can be assessed by 3D ultrasound, therefore diagnosing anomalies of the genitalia is a real possibility.[31] It is possible to asses the volume of the urinary bladder and to evaluate better some anomalies like bladder extrophy.[32] However there are reports suggesting only a small improvement in diagnosing genito-urinary anomalies by 3D US.[13,33] In our study genito-urinary system abnormalities advantageously evaluated in 17 of 31 anomalies *(Figure 6)*.

Three-dimensional (3D) power Doppler sonographic imaging provides a 3D view of the blood vessels.[34] This technique reportedly has advantages over other forms of sonography in visualizing normal and abnormal fetal vascular anatomy.[34,35] Additionally vascular structures of structural defects and tumoral pathologies in the fetus can be evaluated entirely with glass body appereance and 3D color or Power Doppler angiography mode very clearly.[4,33,34,36]

Spatio-temporal image correlation (STIC) is a new approach for the clinical assessment of the cardiac malformations. It offers an easy to use technique to acquire data from the fetal heart and to aid in visualization with both two dimensional and three dimensional cine sequences.[37] Once the volume data is processed and displayed, examination of cardiovascular structures can be accomplished by rotating 2D images in an infinite number of planes. In addition 3D surface rendered anatomy of the heart and great vessels can be accomplished. It has a potential to shorten the evaluation time, especially when the complex heart defects are suspected[37–38]. 3D US gives a new perspective to fetal heart scanning especially in cardiac defects.[37–39] We evaluated 15 (62.5%) anomalies with advantageous scanning by 3D US out of 24 anomalies.

Conclusion

Three-dimensional sonography (3D US) has been demonstrated to be a promising technique for detecting fetal malformations. It is now clear that in the near future 3D technology will be present on all ultrasound machines. 3D US can be a powerful adjunctive tool to 2D US in providing a more comprehensible 3D US impression of congenital anomalies. Although there are still some problems like surface rendering in oligohydramnios and movement artifacts during volume acquisition that need to be resolved, three-dimensional ultrasound opens-up a new dimension in ultrasound investigation.

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