



ULTRASOUND DIAGNOSIS OF CONOTRUNCAL HEART DEFECTS

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Abstract. We present some cases of fetuses with conotruncal heart anomalies (CTA) diagnosed by four dimensional ultrasound (4D US). Improvement of detection and complete prenatal evaluation of conotruncal heart defects is possible if 4D US and STIC technique are available [1]. Fetal echocardiography was done with a 4D ultrasound system (Voluson E8, GE Healthcare, Kretztechnik, Zipf, Austria) equipped with a 4–8 MHz transabdominal transducer and STIC. All pregnant women underwent comprehensive evaluations with STIC, these views were stored and then shared for expert review, interdisciplinary consultation and parental counseling [2]. 4D-STIC echocardiography carried out by experts in fetal echocardiography can be used to accurately diagnose CTA.

Keywords: conotruncal heart defects, prenatal diagnosis, echocardiography, 4D ultrasound, spatio-temporal image correlation

Introduction

Congenital heart disease (CHD) is the most common severe congenital abnormality with an incidence of 8 to 9 per 1000 live births [3]. The majority of fetuses with CHD have no known risk factors. In most clinical settings, only a small percentage of isolated major heart defects are detected prenatally, resulting in worse neonatal and infant outcome. Conotruncal malformations (tetralogy of Fallot, double outlet right ventricle, truncus arteriosus, complete transposition of great arteries, corrected transposition of great arteries and interrupted aortic arch) are the leading cause of symptomatic cyanotic heart disease in the first year of life, accounting for 20-30% of all cardiac anomalies [4]. These malformations may be associated with deletion of chromosome 22q11.2 (Di George syndrome diagnosed by FISH analysis). Nowadays the recognition of these anomalies remains difficult. The four chamber view is frequently unremarkable

[5]. Over the last few years, three-dimensional (3D) and 4D facilities have been used in fetal echocardiography, especially with the advent of the exciting acquisition technique related to a beating fetal heart, called spatio-temporal image correlation (STIC). STIC software is an acquisition modality for the cardiac volume, indirect, motion-gated, off-line, correlating multiples images from different heart cycles. The quality of information depends on the quality of the two-dimensional (2D) image, the frame rate, the angle sweep and the acquisition time. It could be combined with other applications (B-flow, color and power Doppler, tissue Doppler, high-definition flow Doppler) or with postprocessing visualization modalities (surface mode display, tomographic ultrasound imaging, inversion mode, transparent minimum mode) [6]. The reference plan is the four chamber plane with minimal shadowing by the fetal skeleton. STIC has the potential to shorten the evaluation time of complex heart defects; data volume can be stored and reviewed offline by experts and could help to explain fetal anomalies to the parents. It also presents gray-scale or color Doppler information or both for diastole and systole. The major limitation of the STIC is in the advanced gestational age, the increased

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mineralization of the ribs and the sternum and the necessity of having a volume that includes the whole fetal thorax, from the transverse abdominal view to the upper mediastinum [7].

Objectives

STIC is the modern method for establishing fetal heart anatomy; there were several papers to check it as a tool for screening the normal heart. However, its usefulness in cases of heart defects is not well described yet. We aimed to determine whether 3D/4D US with STIC improved diagnostic ability of conotruncal anomalies (CTA) in fetal heart scanning.

Material and method

This is a prospective ongoing study (September 2008-October 2010) on 300 women who booked in for early or mid-trimester targeted organs scan that had a complete fetal echocardiography according to our 5 planes protocol plus the ductus venosus and longitudinal aortic arch planes, performed with 2D US, 2D color Doppler, STIC, STIC with color Doppler, and STIC with B-flow. Ultrasound examination was performed using a Voluson E8 Expert (GE Healthcare) equipped with a 4-8 Mhz probe and for offline analysis we used 4D view software. Cases with CHD were stored in a dedicated archive. The volume data were analyzed with 4DView 2000 software (General Electric Medical Systems, Kretztechnik).

Diagnosis was confirmed by pathological examination or neonatal echocardiography. Subjects were mixed, low and high-risk patients presenting for obstetric scanning between 20 and 32 weeks of gestation. We first acquired a 2D US 30° sweep of the fetal abdomen to the upper mediastinum. The STIC volumes were acquired with acquisition time and angle ranging from 10/25° seconds to 12,5/40° seconds, as appropriate. Karyotyping and FISH for microdeletion 22q11.2 were performed in all cases. The standardized acquisition plane was the four chamber view, preferably by an apical approach. All volumes were acquired using speckle reduction imaging and CrossXBeam CRI (Compound Resolution Imaging). We opened the acquisition box wide enough to ensure that the fetal chest is contained within the box, and we used an acquisition angle that is wide enough to include the stomach inferiorly and the lower neck superiorly. The color Doppler maximal velocity setting was adjusted so that the great arteries were homogenous in color, did not demonstrate aliasing and the color signal filled the lumen of the vessels.

Results

The median gestational age at volume acquisition was 22+4 weeks.

The maternal age ranged from 25 to 39 (mean, 31) years. The incidence of cardiac heart disease (CHD) was 8%. 4D STIC volume acquisition was possible in all cases. 6 conotruncal anomalies (25%) were determined in the 24 congenital heart defects diagnosed at postnatal evaluation: 2 complex Fallot, 2 transpositions of great arteries, one double outlet ventricle, one interruption of aortic arch type B. One patient had known risk factors: in the case of double outlet right ventricle, there was a familial history of cardiac disease. STIC echocardiography identified all cases and had added value in one case (16.6%) - right aortic arch with coarctation and anomalous branching diagnosed with B-flow. Chromosomal malformation affected 2/6 fetuses in which a karyotype was available.

Case 1. A 30-year-old-woman presented to the outpatient clinic for routine ultrasound exam at 23 GW. The first trimester ultrasound screening was normal and there was no exposure to drugs or toxins or history of cardiac anomalies in her medical records. The four chamber view was normal (figure 1), but the aortic artery overrode the interventricular septum (figure 2). After STIC was performed, looking at the great vessels, we noticed the stenotic pulmonary artery in a single view of multiple planes and no other cardiac anomalies. Extracardiac ultrasound exam was normal. Diagnosis was tetralogy of Fallot. Amniocentesis revealed Down syndrome. The parents decided on interrupting the pregnancy and the diagnosis was confirmed at necropsy (figure 3).



Figure 1. Four chamber view

Case 2. A 27-year-old multiparous woman was referred to our unit because of suspected cardiac abnormalities at 21 weeks of gestation. Two-dimensional (2D) echocardiography showed two parallel great vessels arising from the right ventricle in the outflow-tract view and the diagnosis of DORV was made. Color Doppler flow confirmed it. STIC

revealed great vessels arising in parallel from the right ventricle. The aorta and the pulmonary artery were seen to come in parallel from the right ventricle, whereas both these two vessels cross in a normal heart. Therefore the diagnosis of DORV was confirmed. The parents decided on interrupting the pregnancy and the diagnosis was confirmed at necropsy.



Figure 2. Artery override the interventricular septum



Figure 3. Necropsy confirmed diagnosis

Case 3. A 32-year-old woman gravida 2 para 1, was referred to our unit at 22 gestational weeks (GW) after routine dating ultrasound, that revealed the heart anomaly. On the four chamber view, a disproportion of the ventricles was seen. After STIC acquisition we were able to see a straight course of the aorta from the heart to the neck. With B flow diagnosis, interrupted aortic arch type B was confirmed. The parents decided on pregnancy termination after refused FISH analysis for detection of microdeletion of chromosome 22q11.2.

Case 4. A 23-year-old primigravida, 22 GW, was referred to our unit for second trimester ultrasound screening. There was no history of cardiac anomalies or exposure to drugs or toxins in her medical records. The ultrasound examination showed a single fetus with biometric measurements

corresponding to 22 GW. The four-chamber view showed a normal heart with balanced chambers, normal atrioventricular connection, intact crux and intact interventricular septum. A straight course arterial vessel arose from the left ventricle with lateral branches (figure 4). At the level of the 3VT view, two vessels rather than three, (transverse aortic arch and superior vena cava), were seen in this case. STIC with TUI revealed important information on the relationship of the great vessels. Because we suspected TGA, we moved the transducer from the four-chamber view toward the upper thorax 4–6-mm, then tilted the probe 25–50° to demonstrate that the great vessels were parallel (figure 5). In cases of TGA, the en face view of the great arteries and atrioventricular valves resembles a famous Japanese fictional character, ‘Keroppi’ and this is known as the ‘big-eyed frog’ sign. The parents decided on pregnancy termination.



Figure 4



Figure 5

Discussions

The *tetralogy of Fallot (TOF)* is the single most common malformation in children born with cyanotic heart disease. It is characterized by subaortic ventricular septal defect (VSD), and aortic root that overrides the VSD and infundibular pulmonary stenosis. Because of variant shunts that exist in

the fetal circulation, the right ventricular hypertrophy may not be present in utero [13]. The four chamber view appears normal, unless the VSD is large and visible in this plane. The aortic override is due to a discontinuity between the interventricular septum and the medial aortic wall, a situation named aortic dextroposition. The degree of right ventricular outflow tract obstruction is thought to be the significant factor in determining the type and amount of dysfunction that a patient with tetralogy of Fallot displays [8]. The five-chamber view may also be useful when using pulsed Doppler or color Doppler imaging to appraise aortic insufficiency that may result from root dilatation. STIC applied to a three dimensional volume obtained at the level of the four chamber view enables the demonstration of the VSD, aortic overriding, and the stenotic pulmonary artery in a single view of multiple planes. STIC with color Doppler revealed in a glass body mode, a clear demonstration of the lesion in the three vessel trachea view. When diagnosis is confirmed postnatally, the baby undergoes cardiac catheterization to identify collateral vessels in addition to estimate pulmonary and coronary anatomy. The definitive treatment of this heart lesion is surgical, although medical management before and after surgery is important. The prognosis for patients with TOF is variable and is primarily dependent on the pulmonary stenosis.

Double outlet right ventricle: is a type of ventriculoarterial connection in which both great vessels arise either entirely or predominantly from the right ventricle. The diagnosis is made by observation of the pulmonary artery and aorta linked to the right ventricle. Most often the aorta is side by side to the pulmonary artery right lateral, in a long axis view [9]. The great vessels seem to travel a parallel course. The four chamber view is normal, the five chamber view is abnormal as it shows the VSD. There is a lack of continuity of the medial wall of the aorta with the ventricular septum, and the origin of both great arteries comes from the anterior chamber. The VSD often has a subaortic location. Color Doppler assists in visualizing the blood flow from the right ventricle into both the pulmonary artery and the aorta. STIC can help present the type of spatial arrangements of the great arteries and the anatomic relationship of the VSD to the great arteries. Pulmonary stenosis is in many cases an associated anomaly. This malformation requires surgical intervention. The initial management is medical with prostaglandin E until the time of surgical intervention. Surgical repair is taken on to perform a complete two-ventricle repair in order to restore normal circulation. The prognosis depends on associated anomalies. The survival rate is 38%.

Complete transposition of great arteries. In this

malformation, the aorta is connected to the right ventricle and the pulmonary artery to the left ventricle and there is a complete atrioventricular concordance. Both great arteries display a parallel course, with the aorta anterior and to the right of the pulmonary artery. Ultrasound diagnosis shows a normal four chamber view [10]. From the subcostal four chamber view we obtain the outflow tracts. The great vessels are parallel, not crossing at the level of the semilunar valves. Visualization of the five chamber view shows the pulmonary artery arising from the left ventricle and bifurcating, shortly after its origin, into two branches: the right and left pulmonary arteries; the aorta is anterior and parallel to the pulmonary artery. The large vessel noted in the three vessel trachea view is the aorta. Color Doppler aids in demonstrating the parallel course of the great vessels and the associated VSD. STIC evaluation demonstrates the abnormal feature of ventricular-arterial connection in all fetuses and predicts the possibility of abnormal coronary arterial distribution. VSD is the most commonly associated anomaly. Two major surgical techniques have been put into practice in this disorder: the atrial switch procedure and the arterial switch procedure. The survival rate is relatively good.

Congenitally corrected transposition of the great arteries is a rare cardiac anomaly described by atrioventricular and ventriculoarterial discordance. The morphologic right atrium is connected to the morphologic left ventricle by the mitral valve, and the morphologic left atrium is connected to the morphologic right ventricle by the tricuspid valve. The great vessels are also relocated and discordantly connected to the ventricles. The ultrasound diagnosis is established upon the recognition of atrioventricular discordance. The left atrial appendage is finger-like and long, while on the contrary the right atrial appendage is broad based. The ventricles are identified by the atrioventricular valve and musculature. In the four chamber view we see the typical ventricular morphology. Color Doppler is important for the detection or exclusion of prevalently associated intracardiac abnormalities and helps in revealing the parallel course of the great vessels. STIC can be of help in demonstrating the anatomy of the ventricles and the atrioventricular valves in addition to the origin and course of the great vessels. The prognosis of this anomaly is good, aside from its association with other anomalies such as severe tricuspid regurgitation or atrioventricular block [11].

The *interruption of the aortic arch* is characterized by complete separation of the descending and ascending aorta. There are three types of interruption of the aortic arch, according to the level of interruption compared to the brachiocephalic

vessels. In type A (42%), the pulmonary artery supplies the descending aorta via the ductus, and the aorta supplies the three brachiocephalic vessels. The interruption is proximal to the left subclavian artery in type B (53%). In type C (4%), the interruption is between the left common carotid artery and the right innominate. Type C is the most severe form. Ventricular septal defects occur in 50% of type A and are always present in type B. In the three-vessels-trachea view the continuity of the aortic arch cannot be revealed. Ventricular septal defect could be verified by color Doppler in the four and five-chamber view. The diagnosis is confirmed by the straight course of the aorta from the heart to the neck. STIC in combination with B flow in the rendered mode can present the straight course of the interrupted arch. Neonates with interrupted aortic arch develop acidosis and irreversible heart failure; the disease could be corrected when diagnosis is made early [12].

Conclusion

4D-STIC echocardiography carried out by experts in fetal echocardiography can be used to accurately diagnose CTA. STIC with TUI show significant data on the spatial relationship of the aorta and the pulmonary artery in fetuses with TGA. With respect to 2D ultrasound, 4D fetal sonography with STIC and B-flow imaging are able to visualize the anatomical features of IAA and its different types, supplying additional information.

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References

1. DeVore GR, Falkensammer P, Sklansky MS, Platt LD. Spatiotemporal image correlation (STIC): new technology for evaluation of the fetal heart. *Ultrasound Obstet Gynecol* 2003;22: 380–387.
2. Goncalves LF, Lee W, Chaiworapongsa T, Espinoza J, Schoen ML, Falkensammer P, Treadwell M, Romero R. Four dimensional ultrasonography of the fetal heart with spatiotemporal image correlation. *Am J Obstet Gynecol* 2003; 189:1792–1802.
3. Falkensammer P. Spatio-temporal image correlation for volume ultrasound. Studies of the fetal heart. *GE Healthcare: Zipf, Austria*, 2005.
4. Goncalves LF, Lee W, Espinoza J, Romero R. Examination of the fetal heart by four-dimensional (4D) ultrasound with spatiotemporal image correlation (STIC). *Ultrasound Obstet Gynecol* 2006; 27: 336–348.
5. Yagel S, Cohen SM, Achiron R. Examination of the fetal heart by five short-axis views: A proposed screening method for comprehensive cardiac evaluation. *Ultrasound Obstet Gynecol* 2001; 17: 367–369.
6. Yagel S, Arbel R, Anteby EY, Raveh D, Achiron R. The three vessels and trachea view (3VT) in fetal cardiac scanning. *Ultrasound Obstet Gynecol* 2002; 20: 340–345.
7. DeVore GR, Polanco B, Sklansky MS, Platt LD. The ‘spin’ technique: a new method for examination of the fetal outflow tracts using three-dimensional ultrasound. *Ultrasound Obstet Gynecol* 2004; 24: 72–82.
8. Abuhamad A. Automated multiplanar imaging: a novel approach to ultrasonography. *J Ultrasound Med* 2004; 23:573–576.
9. Espinoza J, Kusanovic JP, Goncalves LF, Nien JK, Hassan S, Lee W, Romero R. A novel algorithm for comprehensive fetal echocardiography using 4-dimensional ultrasonography and tomographic imaging. *J Ultrasound Med* 2006; 25: 947–956.
10. Yagel S, Benachi A, Bonnet D, Dumez Y, Hochner-Celnikier D, Cohen SM, Valsky DV, Fermont L. Rendering in fetal cardiac scanning: the intracardiac septa and the coronal atrioventricular valve planes. *Ultrasound Obstet Gynecol* 2006; 28: 266–274.
11. Goncalves LF, Romero R, Espinoza J, Lee W, Treadwell M, Chintala K, Brandl H, Chaiworapongsa T. Four-dimensional ultrasonography of the fetal heart using color Doppler spatiotemporal image correlation. *J Ultrasound Med* 2004; 23:473–481.
12. Chaoui R, Hoffmann J, Heling KS. Three-dimensional (3D) and 4D color Doppler fetal echocardiography using spatio-temporal image correlation (STIC). *Ultrasound Obstet Gynecol* 2004;23: 535–545.
13. Wisniewski KB. *Tetralogy of Fallot*. Drose JA, eds. Fetal echocardiography. Philadelphia, Pa: Saunders, 1998; 185-194.