How soon can fetal echocardiography be offered in cases of suspected CHD at first trimester scan and how accurately can it be achieved?

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Objective
To compare the cardiac findings at 15-16 weeks’ gestation with the 20-22 weeks evaluation in suspected cases of congenital heart disease (CHD) at first trimester screening and to determine the accuracy of early second trimester echocardiography.

Methods
We searched from our database for all cases where a CHD was suspected at the first trimester screening for aneuploidies from 2010 to 2014. All patients underwent fetal echocardiography at 15-16 weeks and were reviewed at 20-22 weeks’ gestation. In all of the antenatal cases of congenital heart disease, the diagnosis was established conclusively by postnatal echocardiography, surgery or postmortem evaluation.

Results
120 cases of CHD were suspected at 11-14 weeks during the period of our study. Among them, 46 cases were excluded: in 41/46, parents opted for termination of pregnancy and 5 cases were lost at follow up. Four out the remaining 74 fetuses were defined as normal at both 16 weeks and 20-22 weeks echocardiography. In 66/70 cases the same cardiac abnormality was seen at both cardiac evaluations. Four diagnoses were discordant between the 16 weeks scan and the 20-22 weeks fetal echocardiography (5, 5%); particularly in 2 cases the discordance was due to morphological progression of the cardiac malformation: 1 tricuspid regurgitation (TR) at 15-16 weeks progressed to pulmonary stenosis at 22 weeks; 1 case of tetralogy of Fallot (TOF) was diagnosed at 21 weeks in another fetus with a malaligned ventricular septal defect (VSD) detected at the early scan. Furthermore in 2 fetuses there was a misinterpretation of the fetal defect: one case of double outlet right ventricle (DORV) at 22 weeks was otherwise defined as transposition of the great arteries (TGA) at the early cardiac examination; one VSD at 15 weeks was diagnosed as a partial atrio-ventricular septal defect (AVSD) at 21 weeks. Therefore the accuracy of 15-16 weeks’ fetal echocardiography when compared to 20-22 weeks evaluation was 94.5%, with sensitivity of 94.3% and 100% specificity.

Conclusion
An high degree of accuracy in the identification of CHD can be achieved by fetal echocardiography at 15-16 weeks. A small number of cases showed progression of findings until the 22nd weeks’ gestation, which is consistent with the natural history of the obstructive congenital heart defects throughout the pregnancy. In cases of suspected CHD at the first trimester scan, fetal echocardiography at 15-16 weeks can be offered to identify the cardiac defect with high feasibility and accuracy.