First trimester diagnostic accuracy of two-dimensional ultrasound technique in congenital heart diseases and great arteries anomalies

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Objective
To assess the accuracy of a standard first-trimester (FT) conventional two-dimensional ultrasound (2DUS) examination protocol in detecting congenital heart diseases (CHDs) and great arteries anomalies, in an unselected population.

Methods
This is a 6 years single center study, performed in a FT screening for aneuploidies program. We stored the cardiac sweep digital video clips, in duplex two-dimensional/two dimensional color, from an oblique lateral insonation from the right shoulder. We used 2DUS re-examination by a team of specialists, pathological examination, and subsequent re-examination as the reference standard methods.

Results
In 13810 FT scanned fetuses, positive 2DUS diagnosed both major and minor CHDs with high accuracy (specificity 99. 9%). Positive likelihood ratios were 1896. 83 for major and 920 for minor heart anomalies. Sensitivity was lower for minor defects than for major CHDs (40% vs 55. 17%) with discrepancies between positive predictive values (40% vs 88. 89%). The detection rates varied widely being: 91% (10/11) for complete atrioventricular septal defect (cAVSD), 83% (5/6) for hypoplastic left heart syndrome (HLHS), 80% (4/5) for isolated great arteries transposition (GAT), 25% (3/12) for tetralogy of Fallot (TOF) and 16. 6% (1/6) for isolated aortic coarctation (AoCo).

Conclusion
In late FT the 2D ultrasound is an accurate tool in screening for some of the major CHDs. Using a standardized and low time-consuming technique will probably raise detection rates in this specific group of diseases. It will lower the operator-dependency and will eliminate the fetal position-dependency, two main reasons for the delayed diagnosis in certain CHDs cases. Still, due to the spectrum bias, with poor FT ultrasound markers in some cases and the evolving pattern in others, the overall accuracy of the method is too low for introducing it in national screening programs.