First trimester 2D cardiac assessment for congenital heart diseases

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Objective

To evaluate the accuracy of first-trimester (FT) conventional two-dimensional ultrasound (2D US) examination in detecting major and minor congenital heart diseases (CHD) in a low risk population.

Methods

Our prospective study consecutively enrolled pregnancies referred for nuchal translucency scan, with crown–rump length (CRL) 60–84 mm. We used 4-8 MHz convex transabdominal high-frequency transducers. Digital clips of each fetus were obtained and stored from the initial scan for offline analysis and the color-flow mapping was applied (using a standard fetal protection protocol). We used 2D re-examination by a team of specialists, pathological examination, and subsequent re-examination (second trimester and postpartum) as the reference standard methods.

Results

3240 fetuses (median CRL 68 mm) were examined during the study period. Postnatal confirmation was obtained in 2908 cases (lost of follow-up rate 1. 24%). Out of the 24 fetuses screened positive as major anomaly on 2D US, 21 cases were confirmed. From 7 cases screen positive as minor anomaly, only 4 were confirmed. We had 3 false-positive cases for each minor and major CHDs. Positive 2D US diagnose both major and minor CHDs with high accuracy (specificity 99. 9%). Positive likelihood ratios were 775. 92 for major and 321. 78 for minor heart anomalies. Sensitivity was lower for minor defects than for major CHDs (33. 3% vs 8. 8%) with discrepancies between positive predictive values (57. 14% vs 87. 5%).

Conclusion

With nowadays systems' resolution, first trimester 2D US is a highly accurate tool for CHDs and great arteries anomalies diagnosis prior to 14 weeks of gestation. If results will be replicated by larger prospective studies the issue of elaborating a first trimester screening protocol for CHDs will be raised. It will be probably focused on anomalies that are not satisfactorily reparable and can lead to serious disability.