19th World Congress in Fetal Medicine

Sensitivity of first trimester biochemical and biophysical multiple markers in fetal aneuploidy detection

Azjargal. B1, Urjindelger. TS2, Erdenetuya. G3, Munkhtsetseg. J1 1Department of Biochemistry, school of 1Biomedicine, Mongolian National University of Medical Sciences, 2National Center for Maternal and Child Health 3Department of Pediatrics, School of Medicine, , Mongolian National University of Medical Sciences Mongolian National University of Medical Sciences, S. Zorig Street, Sukhbaatar District, Ulaanbaatar 14210, Mongolia., Mongolia

Objective

To evaluate the efficacy of first-trimester ultrasound and serum markers in fetal aneuploidy detection.

Methods

There were entirely 3854 fetuses with 11-13^{+6} weeks gestation (crown-rump-length- between 45-84 mm) enrolled to assess serum pregnancy-associated plasma protein-A (PAPP-A), betta chorion gonadotropin (b-hCG), nuchal translucency thickness (NT), nasal bone (NB), tricuspid regurgitation (TR) and ductus venosus (DV) flow. The performance of each single and multiple markers for major fetal aneuploidy screening were determined.

Results

Totally, 103 (12.4%) underwent invasive prenatal diagnosis with 15 major chromosome abnormalities identified including 13 cases of 21, 1 case of trisomy 18, and 1 case of 13. NT was the most accurate single marker with a sensitivity of 71.0% and a false-positive rate (FPR) of 4.9 % while DV or TR was the most specific marker (95.2%) but lacked sensitivity. Among multiple first trimester-screening ultrasound markers, NT plus DV evaluation was the most sensitive test (78.5%) with an FPR of 4.76%.

Conclusion

NT was the most accurate first-trimester screening marker for fetal aneuploidy. NT plus DV assessment as double-screening markers could improve the sensitivity by 9% leading to a lower number of unnecessary invasive prenatal diagnoses.