2nd World Congress on
FETAL AND MATERNAL MEDICINE
July 24-25, 2019 Kyoto, Japan

Sensitivity and specificity of integrated test for detection of fetal chromosomal abnormality at 11-13+6 weeks of pregnancy

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Introduction & Aim: Chromosomal anomalies/aneuploidy (such as trisomy 21/down syndrome) is one of the leading causes of perinatal mortality and developmental abnormality. This study was designed to determine the diagnostic accuracy of non-invasive integrated approach (maternal age and serum biochemistry+fetal USG) in detecting fetal chromosomal abnormality in first trimester of pregnancy among Bangladeshi women.

Methods: This prospective observational study was conducted at Maternal-Fetal Medicine (MFM) Obstetrics OPD, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka. Total 76 women at 11-13+6 weeks, carrying singleton fetus were selected. Integrated non-invasive screening methods using combination of Maternal Age (MA), free β-hCG, PAPP-A and fetal USG based measurement of Nuchal Translucency (NT)+Nasal Bone (NB)+Tricuspid Regurgitation (TR)+ductus venosus flow were performed. Risk assessment for aneuploidy was done by using feto-maternal module of the Prisca software (version 5.0) according to the FMF (London) algorithm. Cut-off risk value of 1 in 300 was considered. Data was analyzed by computer with the help of SPSS 17.0.

Result: 5% women become screen positive and 95% were screen negative. Three chromosomal abnormalities were identified including two cases of trisomy 21 and single case of Turner syndrome. The sensitivity and false negative rates for detection of trisomy 21 was 100% and 0% respectively. And overall, sensitivity, false negative rates, specificity and false positive rates of the integrated risk assessment tool for detection of all aneuploidy was 66.7%, 33.3%, 98.6% and 1.4%, respectively.

Conclusion: Non-invasive integrated screening method can detect aneuploidy up to 100% while reducing the false-positive rate by 0%, may lead to decreased number of invasive tests.

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