The value of combined application of multiple prenatal diagnostic techniques in the prenatal diagnosis of fetal chromosomal abnormalities

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Objective: To explore the value of combined application of multiple prenatal diagnosis techniques in prenatal diagnosis of fetal chromosomal abnormalities.

Methods: A retrospective analysis was conducted on 5692 pregnant women who underwent amniotic fluid karyotype analysis combined with bacterial artificial chromosomes-on-beads (BoBs) labeling technique or chromosomal microarray analysis (CMA) technique from January 2020 to June 2023. Results: Abnormal detection rate is 13.91%. Among them, There was no significant difference between the two groups (P>0.05). Among the 792 cases of abnormal chromosomes, karyotype analysis alone detected chromosomal abnormalities mainly in the form of balanced rearrangements and mosaicism. Compared with karyotype analysis, BoBs and CMA detected an additional 266 cases of copy number variations (CNVs), including 64 cases of pathogenic and possibly pathogenic CNVs, and 202 cases of VOUS and possibly benign CNVs. In the group of older pregnant women, the detection of chromosomal abnormalities was mainly in the form of numerical abnormalities, and there was a significant difference in the detection rates of numerical abnormalities and CNVs (P<0.05). In the group with abnormal ultrasound findings, the detection of chromosomal abnormalities was mainly in the form of CNVs, and there was a significant difference in the detection rates of CNVs and numerical abnormalities (P<0.05). There was no significant difference in the detection rates of chromosomal numerical abnormalities and CNVs between the high-risk group of serological screening and the high-risk group of noninvasive prenatal genetic testing (NIPT) (both P>0.05).Conclusion: karyotype analysis combined with BoBs/CMA detection should be performed simultaneously to fully utilize the complementary advantages of cytogenetics and molecular genetics prenatal diagnosis techniques, thereby effectively reducing the incidence of birth defects.

Keywords: Multiple Prenatal ,Prenatal Diagnosis,Fetal Chromosomal Abnormalities

Themes: Newborn health/Fetal health care/perinatal care

Biography

Liu Yu has over 10 years of experience in clinical medical practice and teaching in obstetrics and gynecology. She holds certifications in cytogenetics, prenatal screening, and genetic counseling. Liu Yu specializes in perinatal healthcare, prenatal screening, genetic counseling, managing pregnancy complications, and treating gynecological endocrine disorders. Skilled in research and analysis, she has led scientific research projects and published numerous papers in CSCD-indexed journals.