PRENATAL BACS-ON-BEADS IN THE DETECTION OF COMMON CHROMOSOMAL ABNORMALITIES AND MICRODELETIONS: AN EXPERIENCE FROM VIETNAM

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ABSTRACTS

In prenatal diagnosis, karyotyping is considered the gold standard in detecting chromosomal abnormalities. Despite the long turn-around time of 2-3 weeks due to the need of culturing, this technique gives a detailed visual chromosomal representation. In recent years, implementations of molecular techniques not only eliminates the need for cell culture, which shortens the turn-around time to within 24 hours, but also deliver a similar diagnostic results with a more targeted approach. Besides capillary electrophoresis (QF-PCR) and microarrays, BACs-on-Beads, specifically Prenatal BoBs, has been extensively exploited for their high throughput and multiplex capabilities. In addition to the common abnormalities in chromosome 13, 18, 21 and sex chromosomes, Prenatal BoBs simultaneously provide the detections of 9 most common severe microdeletions which cannot be detected by karyotyping. These include DiGeorge, Williams-Beuren, Prader-Willi, Angelman, Smith-Magenis, Wolf-Hirschhorn, Cri du Chat, Langer-Giedion, and Miller-Dieker.

OBJECTIVES

To evaluate the capability and feasibility of Prenatal BoBs as a routine procedure in prenatal diagnosis

METHODS

Amniotic fluid samples at gestation age of 17-26 weeks were taken from 232 high-risk pregnancies (maternal age of 35 years and over, abnormal ultrasound results, and high-risk evaluation of biochemical assessments). Both conventional karyotyping and Prenatal BoBs were performed on the collected specimens.

RESULTS

All samples returned results for both karyotyping and Prenatal BoBs assays. We did not find false-positive results for Prenatal BoBs. The failure rate was 0% and the overall abnormality detection rate was 6.93% (17 out of 232 amniotic samples were reported abnormal).

CONCLUSIONS

Prenatal BACs-on-Beads assay provides accurate detections of common aneuploidies of chromosome 13, 18, 21 and sex chromosomes (X,Y) with additional 9 common severe microdeletions within 24 hours, thus represents a suitable routine diagnostic procedures. However, Prenatal BoBs cannot replace the importance of visual diagnosis provided by conventional karyotyping.