Rapid molecular karyotyping for products of conception by BACs-on-Beads technology

Objective: Conventional karyotyping is the gold standard for prenatal diagnosis but in products of conception (POC) specimen which requires for tissue culture process, takes a long time and likely high failure rate. Bacterial artificial chromosomes (BACs)-on-Beads (BoBs) technology is a molecular genetic test with do not require cell culture. The objective of this study was to investigate the effectiveness and evaluate the use of BoBs technology for the rapid screening of POC.

Method: We evaluated 20 archived DNA samples collected from products of conception which included samples extracted from placental villi or fetal somatic tissue to detected chromosomal abnormalities. The KaryoLite(TM) BoBs was used in 18 samples and the remaining two samples were used both of KaryoLite(TM) and Prenatal(TM) BoBs kits.

Result: Our BoBs results demonstrated that normal karyotype were detected in 11 samples with approximately equal male/female ratios (55%) and showed the detection of chromosomal abnormalities in 9 samples (45%). The abnormalities were identified as trisomy 13, 16, 20, 21, 22 and monosomy X. The results from 2 samples (1 normal and 1 abnormal) which used both of KaryoLite(TM) and Prenatal(TM) BoBs kits were concordance.

Conclusion: The BACs-on-Beads technology is potentially very useful method for the rapid diagnosis of common aneuploidies and microdeletion syndromes in products of conception because of an accurate, rapid detection and do not require cell culture with the ability to detect in samples that fail in cell culture.