B47 Fetal DNA Detection in Pregnancy Serum Using the Ion Personal Genome Machine® (PGM™) System With an Identity Single Nucleotide Polymorphism (SNP) Panel

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After attending this presentation, attendees will understand the operability of a Next Generation Sequencing (NGS) system in Low Template DNA (LT DNA) analysis, specifically in fetal DNA detection in maternal serum, and the basic principle of the Ion PGM™ platform with the Human Identification (HID) -Ion AmpliSeq™ Identity Panel and its utility in practical casework samples.

This presentation will impact the forensic science community by analyzing DNA obtained from challenging sources, such as degraded DNA and mixed DNA.

NGS systems offer new possibilities in genotype analysis using established forensic markers for identification. The results of the analysis of fetal DNA in maternal plasma or serum is often similar to LT DNA and mixed DNA.

Serum samples were taken from eight anonymous pregnant women. There was no detailed information about the fetus due to the fact that miscarried pregnancies were included and no gender information was available. DNA was isolated using the QIAamp® DNA Mini Kit. DNA quantification was performed using real-time Polymerase Chain Reaction (PCR) and the Quantifiler® Duo Kit. The levels of sequence coverage and mixed DNA patterns were evaluated through HID-Ion AmpliSeq™ Identity Panel with 124-markers designed for the Ion PGM™ system.

Partial Y-chromosome Single Nucleotide Polymorphisms (Y-SNP) markers were typed. Negative Y-STR profiles (no single STR locus) were obtained from serum samples taken from two pregnant women who were assumed to carry male fetuses. Using those particular two Y-SNP samples, partial fetal autosomal SNP profiles were implicitly inferred via retrospective mixture analyses in all of the eight samples.

If additional information, such as portion and degree of mixtures, were available, this platform could be used for non-invasive prenatal testing to detect the mixture samples. This study also suggests that if relevant template chromosomal data are provided, NGS can be readily used to analyze mixed DNA samples. The findings also indicate that NGS will be practical for processing LT DNAs and mixed DNA.

Next Generation Sequencing, Fetal DNA, Mixed DNA