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AB103. A sequence variation of short tandem repeat observed in paternity cases using massively parallel sequencing technology

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Background: STR has been regularly used as genetic marker in paternity testing. The current standard procedure of STR allele detection based on DNA fragment analysis using CE. However, the MPS offers the high-throughput sequencing with lower cost per nucleotide. This technology has increase an opportunity for STR allele detection in national database and paternity testing as well. The high degree polymorphism of STR makes them informative that is an influence of their high mutation rate. Moreover, the usefulness even in homozygous alleles is determined by sequence variation in particular STR marker. In this study, we compare a STR allele designation from CE and a STR sequencing from MPS. The sequence variation has observed in particular STR.

Methods: The DNA of eighteen families were analyzed on a CE using two commercial test kits, while a MPS analysis was performed using MiSeq FG×TM forensic genomics system (Illumina).

Results: The results from MPS platforms showed totally concordant with CE data and also demonstrated the advantage of sequencing-based method in homozygous alleles. The sequencing variation of STR was detected and identified an inherited pattern even on homozygous alleles. For example, locus D3S1358 of child DNA was typed homozygous allele 16,16 on CE while distinguishable sequence suggested $(TCTA)^{1}(TCTG)^{3}(TCTA)^{12}$ from father and $(TCTA)^{1}(TCTG)^{2}(TCTA)^{13}$ from mother, respectively. The variety of STR allele sequences that indicated the allele inherited was detected in 8 of 18 families (44%) in our analysis. The sequence polymorphism was demonstrated in markers, including D2S1338, D3S1358, D4S2408, D5S818, D8S1179, D9S1122 and D21S11. Moreover, the success identification of STR inherited pattern of allele sharing among family also increases a CPI in paternity testing.

Conclusions: The results from MPS platforms showed totally concordant with CE data. The sequence variation of STR from MPS could be indicated the allele inherited pattern in paternity and other relatedness testing.

Keywords: Paternity testing; massively parallel sequencing (MPS); capillary electrophoresis (CE); short tandem repeat (STR); combine paternity index (CPI)

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