VARIANTS OBSERVED FOR STR LOCUS SE33: A CONCORDANCE STUDY

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The degree of concordance of STR allele calls among STR kits is extremely important when comparing STR profiles among laboratories and in national databases. This study focuses on the locus SE33 in recently developed STR kits. The SE33 locus is one of the most polymorphic STR markers used in forensic DNA analyses due to its highly complex repeat pattern motif and high mutation rate. However, the motif complexity can make it difficult to obtain concordant results for some alleles likely due to sequence-dependent conformational changes manifested under different electrophoretic conditions, and/or use of different primers. The AmpFlSTR® NGM SElect™ PCR Amplification Kit (Life Technologies, Foster City, CA), PowerPlex® ESX 17 system (Promega Corporation, Madison, WI), and PowerPlex® ESI 17 system (Promega Corporation) were compared for concordance of allele calls for the SE33 marker. A total of 16 samples were identified that were discordant at one of the SE33 alleles and differed by an apparent one nucleotide in size. The different alleles were observed with the ESI 17 kit while the ESX 17 and NGM SElect kits were concordant. The discordant alleles were observed predominately in individuals of African descent. Sequence analysis revealed that the one-base difference in size was not due to an indel but instead is the result of a single nucleotide polymorphism (SNP) in the flanking region of the SE33 repeat region. Although differences manifested only in the ESI 17 kit, one cannot preclude that similar phenomenon may occur with the other kits as data sets increase. Such differences need to be appreciated in order to ensure that false exclusions do not occur, particularly with database searches, based on a one base pair size difference at the SE33 locus.