STR SEQUENCE DIVERSITY IN POPULATION SAMPLES AND NOMENCLATURE GUIDANCE FOR THE "NEXT GENERATION"

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As STR loci were being identified in the 1990's, various nomenclature systems were developed for different loci, with the primary variation being whether or not to "count" non-repeat bases interspersed in the repeat motif. In 1997, the ISFG issued guidelines on STR nomenclature, in an attempt to provide a common currency for information exchange. Historical precedent already existed for some loci, and this was maintained to avoid confusion, resulting in several commonly used forensic loci having complicated and contradictory nomenclature systems. This has not been an issue within the forensic community, as the capillary electrophoresis (CE)-length analyses are kit-based, with corresponding computer programs that automatically count repeats in a standardized manner. Now, as the costs associated with next-generation sequencing (NGS) methods decline, forensic research laboratories are beginning to explore the increase in information sequencing STR loci may provide. As a new generation of scientists begins interrogating these loci on a deeper level, an understanding of historical nomenclature is needed to achieve bioinformatic concordance with existing CE data. In this presentation, NGS results from population samples will exemplify the sequence variation that exists in forensic STR loci (SNPs and indels within and outside of STR allele regions and repeat motif changes) as well as the complexity and inconsistency of the current nomenclature. This experimental sequence data will give an indication of the level of diversity expected in the larger population and will be used to provide examples of how sub-alleles can improve discrimination and mixture deconvolution in forensic casework. We will discuss the different purposes of nomenclature—manual comparisons, forensic reports, database searching, court explanations—and give examples of possible NGS-compatible nomenclature systems that may meet the needs of the forensic community.