

Hidden Variation in Microsatellite Loci: Utility and Implications for Forensic DNA Analysis

REFERENCE: Planz JV, Hall TA: Hidden variation in microsatellite loci: Utility and implications for forensic DNA analysis; *Forensic Sci Rev* 24:27; 2012.

ABSTRACT: Short tandem repeat (STR) analysis has been the standard in forensic DNA examinations for almost 15 years. The purpose of this article is to provide some perspective on the biological nature of STR alleles themselves, examine underlying distributions of alleles in the STR loci that are routinely used, and to discuss features of these alleles that are not observable with the currently employed methods. Many of the internationally standardized STR loci contain variations of their interrupted repeat structures, either due to the compound or complex nature of the locus or due to nucleotide variations within the simple repeat motif, which inevitably leads them to become more stratified at the population level. Current STR typing procedures utilizing PCR amplification followed by fragment analysis via capillary or gel electrophoresis does not provide the resolution to discern these polymorphisms. Thus, current designation of alleles is operationally and not biologically defined. Although in the comparison of an evidentiary STR profile to that of a potential contributor, the biological nature of the allele may not be of consequence. When comparisons require assumptions of relatedness between individuals, the biological nature of shared alleles becomes an underlying focus. Herein we will discuss the nature of these additional allelic polymorphisms, what is known of their distribution among the STR loci utilized in forensic testing and within populations, and the advantages this level of allelic discrimination has in forensic and relationship testing.

KEY WORDS: Forensics, human identification, mass spectrometry, mutation, population studies, sequence polymorphism, short tandem repeats.
