



B103 Is That Peak Real or Is It a Mis-Type? Only the Profile Donor Knows for Sure

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After attending this presentation, attendees will gain an appreciation for the possibility of frank mis-types in forensic DNA profiles, specifically with regard to peaks in stutter positions of a contributing profile.

This presentation will impact the forensic science community by raising awareness of the possibility for frank mis-types in forensic DNA profiles, especially in low-template samples and in particular, in stutter positions of alleles from a true contributor.

Samples containing low levels of DNA and/or mixtures of DNA from multiple individuals are routinely encountered in forensic DNA casework. These samples can be considered inclusively as “complex” samples. Such samples are challenging to interpret because of the inherent uncertainty in determining the genotypes of the contributors to the evidence profile. To further investigate the challenges involved in interpreting complex samples, a specific sample set was created. This sample set comprised two-, three-, and four-person mixtures. The template amounts were 30pg, 50pg, 100pg, and 500pg. The mixture ratios were 1:1, 2:1, 4:1, and 9:1. The various mixtures resulted in a total of 164 complex samples, each of which was amplified five times for a total of 820 profiles. The sample set provides a rich source of data than can be queried in numerous ways. Because these samples derive from known sources, ground truth is known, and the alleles expected in each of the profiles can be determined.

The Polymerase Chain Reaction (PCR) amplification typing artifact known as stutter introduces an element of ambiguity into the interpretation of forensic DNA profiles. Most analysts are aware of issues that arise from a stutter peak deriving from a relatively high-quantity contributor. These peaks can sometime be higher than expected, introducing the possibility of calling a false allele; they can also potentially mask a real allele in a low-quantity minor component of a mixture; however, there has been little discussion of the behavior of stutter in low-template mixtures and of the potential for the generation of a false profile that does not represent the true profile of the input sample.

In reviewing the aforementioned dataset, it became apparent that a non-trivial proportion of profiles would result in frank mis-types if read blind. Most, but not all, of these instances occurred in low-template samples, and the vast majority of these mis-types were attributed to peaks in either the N-1 or N+1 stutter positions of known input alleles. This phenomenon will be discussed and examples will be shown.

DNA, Mis-Type, Low Template