



B101 Conceptual and Cultural Limitations Delaying the Transition to Probabilistic Genotyping in Forensic DNA Analysis

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After attending this presentation, attendees will better understand the current limitations affecting the transition to probabilistic genotyping in forensic DNA analysis.

This presentation will impact the forensic science community by providing an overview of current practices in Short Tandem Repeat (STR) mixture deconvolution and by contrasting some of these approaches to emerging methods.

The current culture of forensic science teaches us to think in binary (in/out or plus/minus) fashion. Often, the words that are used, including exclude, include, conclusive, inconclusive, match, identity, cannot exclude, etc., are treated as categorical absolutes rather than as proportionate, continuous, or semi-continuous concepts. This commonly accepted usage of terms within the field of forensic science does not serve us well in the real world of uncertainty and continuous variation, especially in cases where the evidentiary data are complex and difficult, if not impossible, to analyze without computer support. Due in part to training, it is difficult to throw away the binary method of thinking and fully embrace continuous reasoning — where certainty is correctly understood as applying within a context rather than absolutely — and uncertainty is embraced and measured rather than avoided.

Most approaches to STR mixture deconvolution have given at least some credence to a quantitative assessment of DNA mixture data, including those advocating thresholds and binary placement into categories; however, as time has passed, the binary approach has slowly been replaced by probabilistic reasoning that attempts to embrace data uncertainty rather than eschew it. So although the germs of these ideas have been present from the outset, they have only recently started to be fully appreciated.

All parties in this discussion agree that quantitative patterns are present in STR-based DNA mixtures, and that the goal of forensic interpretation should be to make full use of these data. Any deliberate restrictions imposed on the data will necessarily result in less than full use of the data. If potentially valuable information is discarded, then potential identification information is also lost, resulting in the potential for the DNA mixture to be considered as uninterpretable, which could result in the guilty remaining free and for cases to remain unresolved. Hence, justice is best served by utilizing the data in the most efficacious manner.

In fully probabilistic approaches to DNA mixture deconvolution, if the data itself are ambiguous, a properly validated computer program will express this ambiguity as uncertainty. This approach does not advocate for deliberate data rejection by applying thresholds that ignore data. If the data are uncertain, the program assigns less probability to particular outcomes, in effect spreading out, or diluting, the resulting probabilities of particular genotypes.

Fully probabilistic genotyping models are not constrained by thresholds and consider all of the genotype probabilities arising from the data. In order to estimate these probabilities, some approaches apply a hierarchical model with many integrated parameter variables, including peak heights, heterozygote peak imbalance, stutter, mixture weights, degradation, and noise.

Variance parameters in DNA mixture deconvolution can most accurately be estimated by using all relevant data, and ignoring relevant data necessarily reduces the accuracy of the estimates. This inevitably leads to higher uncertainty and less-definitive conclusions. Hence, there is no justification for deliberately discarding data, such as in the use of thresholds, nor is there any justification for invoking any “conservative” principles in STR mixture deconvolution, since once the appropriate variance parameters have been identified and modeled, a fully probabilistic approach provides a built-in protection from both overstated and understated conclusions.

A fully rational approach to mixture deconvolution holds that a high value of discrimination is always good. Importantly, efficient mixture deconvolution simultaneously affects both sensitivity and specificity and thus is good for both inclusions (sensitivity), and also for exclusions (specificity). The fully objective view places a great value on a wide separation between these two poles — because reducing uncertainty itself in forensic science is tremendously valuable.

This presentation will provide an overview of the conceptual and cultural limitations within forensic science to the adoption of probabilistic genotyping. Examples will be provided to emphasize the points covered.

DNA Mixtures, Genotyping, Probability