FURTHER EXPLORATION OF TRUEALLELE® CASEWORK

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TrueAllele[®] Casework (TA) is a continuous probabilistic modeling system that utilizes Markov chain Monte Carlo (MCMC) sampling in order to perform statistical modeling of short tandem repeat (STR) data (1,2). Widely ranging disciplines such as physics, psychology, computer learning, economics, biological systems and DNA analysis, utilize probabilistic modeling to make sense of the patterns observed in complex data and predict likely outcomes for various tests (3,4,5). After modeling has been performed by TA, a comparison with reference profiles is performed, likelihood ratios calculated and reported in logarithmic form log(LR)).

After extensive validation work, the Virginia Department of Forensic Science (VDFS) went online for casework analysis with TA in 2014 (6,7). TA is used at VDFS for analysis of complex mixtures comprised of up to four contributors and is used on the most challenging of mixtures tackled by the laboratory. Since implementation, additional validation tests have been performed. These studies examined: 1) What happens to the TA analysis when either a greater or fewer number of contributors is solved for than are actually in the mixture? 2) What effect does the use of the degradation function have on the log(LR) of contributors to a differentially degraded mixture? and 3) What happens to the log(LR) of contributors when the DNA sample is over-amplified?

As reported, when a greater number of contributors is hypothesized for TA analysis than is in the mixture, typically there is a small, if any, effect on the log(LR) values generated (8). When a smaller number of contributors is hypothesized, it can dramatically reduce the log(LR) of donors. This is consistent with how the TA modeling works; restricting the number of contributors also restricts the potential genotype combinations that can explain the data, which may produce a reduction in the log(LR). Providing a greater number of potential contributors does not restrict the genotype combinations.

The use of the degradation function produced only a small (~1 log(LR) unit) change, if any, for contributors in differentially degraded mixtures. However, it may affect how readily the sample is deconvoluted (fewer computer runs). This improvement may be due to a more accurate assessment of the mixture weights when differential degradation is taken into account.

Slightly over-amplified samples produced a small reduction in the log(LR) produced for contributors, but an excessively over-amplified sample could not be successfully analyzed using the same contributor number. This is also consistent with the TA process; over-amplified samples exhibit excessive artifact which increases genotype uncertainty and thus can reduce the log(LR) values produced.

The results of the additional studies were consistent with the earlier work performed at VDFS and what has been reported in the literature for TA.

¹ Perlin MW, Legler MM, Spencer CE, Smith JL, Allan WP, Bellrose, JL, et al. Validating TrueAllele® DNA mixture interpretation. J Forensic Sci 2011 Nov;56:1430-47.

² Perlin MW, Belrose JL, Duceman BW. New York State TrueAllele® Casework validation study. J Forensic Sci 2013 Nov;58(6):1458-66.

³ Richey M. The evolution of Markov chain Monte Carlo methods. American Mathematical Monthly 2010; 117:383-413.

⁴ Poirer DJ. The growth of Bayesian methods in statistics and economics since 1970. Bayesian Analysis 2006;1(4):969-80.

⁵ Gelfand AE, Smith AFM. Sampling-Based Approaches to Calculating Marginal Densities. J American Statistical Assoc 1990;85:398-409.

 ⁶ http://www.dfs.virginia.gov/wp-content/uploads/2014/11/210-D1500-FB-PM-TrueAllele-Casework-System.pdf (accessed July 28, 2015)
⁷ Greenspoon SA., Schiermeier-Wood L., Jenkins BC. <u>Establishing the limits of TrueAllele® Casework: a validation study.</u> Journal of Forensic Sciences, 2015(in press).

⁸ Perlin MW, Hornyak JM, Sugimoto G, Miller KWP. TrueAllele_ Genotype Identification on DNA Mixtures Containing up to Five Unknown ContributorsJ Forensic Sci, 2015 doi: 10.1111/1556-4029.12788