Dr. Mike Coble's NIST April 2013 talk on "Probabilistic Genotyping"

Here are corrections and clarifications to Mike's language and calculations. I included important information (e.g., validation studies) that Mike did not have time to mention.

• TrueAllele got it right

(Slide 28) TrueAllele® Casework objectively infers genotypes from STR data, thoroughly considering all allele pair possibilities. TrueAllele separates mixtures into contributor genotypes using all the peak height data, without being told the suspect's genotype. The computer's inferred genotype is independent of anyone's preferred answer. With data ambiguity, genotype probability is placed on multiple allele pairs, one of which might match a suspect or a known contributor.

• TrueAllele genotype likelihood

(Slide 29) The table shows a *prior* population genotype (HWE) and TrueAllele's computer-inferred evidence *likelihood* values (Prob).

• TrueAllele genotype probability

(Slide 30) The genotype probability is proportional to the product of prior times likelihood (HWE*Pr). Dividing each product by the Total sum 0.0143 normalizes the column to a *posterior* genotype probability. The genotype probability at the suspect's FGA allele pair 20,22 is HWE*Pr/Total, or 0.0080/0.0143, which equals 56%.

• TrueAllele likelihood ratio

The likelihood ratio (LR) is computed at the suspect's row (FGA allele pair 20, 22) by dividing the posterior genotype probability 56% by the prior genotype probability 5.4%, to give a LR of 10.3. The maximum possible LR (e.g., for a single source sample) at this locus would be 100%/5.4%, or 18.5. A genotype probability for the suspect of over half thus preserves most of the identification information.

• TrueAllele genotype listing

The genotype listing shows 9 allele pair rows at a 99.99% cumulative probability level. In the TrueAllele visual user interface (VUIer™), the user controls this probability level, typically set around 95%. A 99% level would show 6 rows, a 95% level only 5 rows, and a 90% level just 4 rows.

• New Zealand population database

(Slide 34) The STRmix operator used a New Zealand native (Māori) enriched population database. Therefore, the LR values in the two examples cannot be directly compared.

• New Zealand genotype probability

STRmix inferred an FGA genotype probability of 24% at allele pair 20,22.

• New Zealand likelihood ratio

Using a US Caucasian population database, the LR at 20,22 would be 24%/5.4%, or 4.4. The very large LR shown (103) results from the relative rarity of FGA genotype 20,22 in New Zealand (very small population denominator), and cannot be meaningfully compared with TrueAllele's US Caucasian statistic (much larger 20,22 population prevalence).

• TrueAllele validation studies

TrueAllele DNA mixture interpretation has been extensively validated. Three peer-reviewed validation papers have been published, one using NIST laboratory data of known composition (PLoS ONE, 2009) and two using casework items (JFS, 2011 & 2013). Other mixture validation studies have been presented at conferences (ISFG, 2011; AAFS, 2013).

• TrueAllele admissibility

TrueAllele Casework has had successful admissibility hearings in Pennsylvania, California and the United Kingdom. TrueAllele has statewide precedent in Pennsylvania, based on favorable appellate rulings.

• TrueAllele reports and trials

Over 100 criminal TrueAllele case reports have been written. TrueAllele testimony has been given in 16 criminal trials. Both the prosecution and defense use TrueAllele to determine accurate LRs from complex DNA mixture evidence.

References

Perlin MW, Sinelnikov A (2009) An information gap in DNA evidence interpretation. PLoS ONE 4: e8327.

Perlin MW, Legler MM, Spencer CE, Smith JL, Allan WP, et al. (2011) Validating TrueAllele® DNA mixture interpretation. J Forensic Sci 56: 1430-1447.

Coble MD, Butler JM (2011) Exploring the capabilities of mixture interpretation using TrueAllele software. International Society of Forensic Genetics. Vienna.

Perlin MW, Dormer K, Hornyak J, Schiermeier-Wood L, Greenspoon S. Virginia TrueAllele® validation study: casework comparison (A125); 2013; Washingon, DC. American Academy of Forensic Sciences. pp. 94.

Perlin MW, Belrose JL, Duceman BW (2013) New York State TrueAllele® Casework validation study. J Forensic Sci 58: in press.