B137 The False Inclusion of Non-Contributors in DNA Mixtures Cases

Marie Semaan, MS, American University of Science and Technology, Beirut, LEBANON; Sarah Abbas, MS, American University of Science and Technology, Beirut, LEBANON; Issam Mansour, PhD, American University of Science and Technology, Beirut, LEBANON*

Learning Overview: The goal of this presentation is to assess the risk of false inclusions when comparing DNA profiles of unrelated persons to simulated mixed traces in Lebanon.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by: (1) shedding light on the effect of allele sharing and homozygosity on the evaluation of DNA mixtures, especially in consanguineous and endogamous communities; (2) shedding light on the risk of false inclusions in DNA mixtures in similar populations; and (3) providing recommendations in similar populations concerning the use of profile-to-mixture matching studies.

The interpretation of forensic DNA mixtures is particularly complex due to several factors, including allele sharing, allele dropout, low copy number, and Polymerase Chain Reaction (PCR) stutter artifacts. Statistical interpretation is essential in reporting DNA results in which various approaches are used to assess the weight of the resulting mixed DNA profile. The present research aimed at assessing the risk of false inclusions when comparing DNA profiles of unrelated persons to simulated mixed traces in Lebanon.

A total of 517 samples were collected from seven Lebanese villages representing different Lebanese regions and religious backgrounds. Profiling was performed with three different profile sizes: 16, 23, and 28 Short Tandem Repeat (STR) systems using three multiplex STR kits: the PowerPlex®16 HS, the PowerPlex® ESI 17, and the PowerPlex® CS7. A specialized in-house software, the DNA Mixture and Matching Simulator, was developed to perform simulation studies, where each individual profile was compared against electronically simulated mixtures of two contributors.

The comparisons of the individual profiles to mixtures of two contributors showed false inclusions with 16-, 24-, and even 29-locus profiles. Sixty-nine non-contributors were falsely included in DNA mixtures with 16 tested loci. With 24 tested loci, 13 non-contributors were falsely included in the mixtures. Five non-contributors were falsely included in DNA mixtures using 29 STR systems. This showed that increasing the number of STR loci increases the power of discrimination and reduces the possibility of false inclusions, but it does not eliminate it.

Physical DNA mixtures of the electronically simulated cases were produced and statistical interpretation was then performed based on the semi-continuous model using three of the most frequently used DNA mixture analysis expert software programs, taking into account the relevant allele Lebanese frequency and inbreeding coefficient of the population, to compute the likelihood ratio. The statistical results of the expert software confirmed the inclusion of non-contributors in the cases with adventitious matches and confirmed these false inclusions.

DNA mixture simulations of three contributors were then performed using the same in-house software where each individual profile was compared against electronically simulated mixtures of three contributors. The comparisons showed 1,100 cases in which a non-contributor was falsely included in a DNA mixture of three contributors using 29 STR systems. Moreover, a challenging case showed eight non-contributors falsely included using 24-locus profiles and five non-contributors falsely included using 29-locus profiles.

These false inclusion cases shed light on the effect of allele sharing and homozygosity on the evaluation of DNA mixtures, especially in consanguineous and endogamous communities. In similar populations, this study recommended using profile-to-mixture matching studies for exclusion purposes only, even when using 29-locus profiles and expert software statistical analysis.

Forensic DNA, Mixed Traces, Expert Software

*Presenting Author