Journal of Forensic Research
ISSN: 2157-7145

Semaan et al., J Forensic Res 2020, 11:2

Case Report Open Access

A Mixed DNA Profile Controversy

Marie Semaan[1], **Sarah Abbas**[1,2] **and Issam Mansour**[1*]

[1]*Department of Laboratory Science and Technology, Faculty of Health Sciences, American University of Science and Technology, Beirut, Lebanon*

[2]*School of Criminal Justice, University of Lausanne, Switzerland*

***Corresponding author:** Issam Mansour, Department of Laboratory Science and Technology, Faculty of Health Sciences, American University of Science and Technology, Beirut, Lebanon, Tel: +961218716230, E-mail: fhs@aust.edu.lb

Received date: February 01, 2020; **Accepted date:** March 10, 2020; **Published date:** March 20, 2020

Copyright: © 2020 Semaan M, et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

Abstract

The DNA trace has been playing a crucial role in identifying or exonerating potential suspects. While single source DNA traces face interpretational challenges, the evaluation of a forensic DNA mixture trace faces much greater challenges especially with increased allele sharing and homozygosity. The present report describes a challenging case where eight potential suspects could not be excluded in a simulated mixed DNA analysis. Even though relevant frequency datasets and an inbreeding coefficient were considered and expert DNA mixture analysis software was used, statistical analysis falsely supported the inclusion of non-contributors. The present case shed the light on the effect of allele sharing and homozygosity on the evaluation of DNA mixtures especially in consanguineous and endogamous populations. Recommendations as to DNA mixture analysis were issued for local forensic uses and for other similar populations.

Keywords: Forensic DNA; Mixed traces; Expert software; Inbreeding; Likelihood ratio

Introduction

Traces involving DNA mixtures are frequently encountered in forensic caseworks [1]. Such traces originate from two or more contributors [2]. Interpretation of mixed traces could ideally be performed when (A) the amount of amplified DNA from all contributors is sufficient and above the analytical threshold; (B) the ratio of DNA contributed by each source is reflected on the peak heights and consequently the possible genotypes of major and minor contributors may be determined [3]; (C) there is no degradation and allele drop out; (D) no artifacts and allele drop in; and (E) the contributors are unrelated and have few shared alleles. But since the situation of forensic cases is rarely ideal and the likelihood in which it is not possible to distinguish the alleles of the different contributors does exist, several models have been suggested for the interpretation of mixed DNA profiles [4].

The present report describes a challenging case where eight different individuals could not be excluded in a mixed DNA analysis. Even though relevant frequency datasets and an inbreeding coefficient were considered and expert DNA mixture analysis software was used.

Case Presentation

The present case is a mixed DNA trace (Figure 1). Profiling was performed with 23 STR systems by combining two multiplex STR kits: PowerPlex® 16 HS, the PowerPlex® ESI 17 (Promega Corporation; Madison, WI, USA). Systems D3S1358, D5S818, TH01, D21S11, TPOX, D7S820, D2S1338, Penta D and D1S1656 (highlighted in blue) and systems D8S1179, SE33 and D12S391 (highlighted in yellow) show that at least two individuals contributed to this trace.

Locus	Trace
D3S1358	15 17 18
D19S433	13 16
D8S1179	8 12 13 15
D5S818	11 13 14
TH01	7 9 10
vWA	14 20
D21S11	29 30 30.2
D13S317	10 12
TPOX	8 10 11
FGA	22 23
D7S820	9 10 12
D16S539	11 12
D18S51	12 19
CSF1PO	11 12
D2S1338	18 19 20
Penta E	5 12
Penta D	8 11 12
SE33	19 25.2 28.2 29.2
D22S1045	11 15
D1S1656	12 16 17
D10S1248	12 14
D2S441	11 14
D12S391	19 21 22 23

Figure 1: Profile of the mixed trace.

Only two alleles per locus appear in systems D19S433, vWA, D13S317, FGA, D16S539, D18S51, CSF1PO, Penta E, D22S1045 and

D2S441 (highlighted in green), which is potentially due to homozygosity and allele sharing [5] characteristics that are frequent in the Lebanese population due increased inbreeding in the community under study [6-8]. Allele drop out could be a further reason to whether only two or more profiles contributed to the mixture obtained from the trace.

Eight potential suspects were considered, based on all other non-DNA evidence relevant to the allegation [9], all of whose DNA profiles showed a complete adventitious match with the mixed DNA trace (Figure 2). Statistical interpretation was performed based on the semi-continuous model using the DNA mixture analysis expert software LRmix studio, for computing the likelihood ratio. The allele frequencies of the Lebanese population were considered as well as the inbreeding coefficient that reflects the rate of inbreeding in the Lebanese population.

The following LR values that ranged between 10^{81} and 10^{94} were obtained (Figure 2). The statistical results confirmed the inclusion of the eight suspects and none of the individuals could be excluded by any of the 23 tested loci.

Locus	Trace	Suspect 1	Suspect 2	Suspect 3	Suspect 4	Suspect 5	Suspect 6	Suspect 7	Suspect 8
D3S1358	15 17 18	17 18	15 17	17 18	15 18	15 17	15 17	15 17	15 17
D19S433	13 16	13 13	13 16	13 16	13 16	13 13	13 16	13 16	13 16
D8S1179	8 12 13 15	12 13	12 15	12 13	8 12	12 15	8 15	8 13	12 15
D5S818	11 13 14	11 14	11 13	13 14	11 13	11 13	11 11	11 14	11 14
TH01	7 9 10	9 9	9 10	7 10	9 10	9 9	9 10	9 10	9 10
vWA	14 20	20 20	14 20	14 20	14 20	14 20	20 20	20 20	14 14
D21S11	29 30 30.2	29 30	29 30	30 30.2	30 30	29 30	29 30	29 30	29 30
D13S317	10 12	10 12	12 12	10 12	10 12	12 12	12 12	12 12	10 12
TPOX	8 10 11	10 11	8 10	8 11	8 10	10 10	8 10	8 11	8 10
FGA	22 23	22 23	22 22	23 23	22 23	22 23	22 23	22 23	22 23
D7S820	9 10 12	10 12	10 10	9 10	9 10	9 12	10 12	10 12	9 10
D16S539	11 12	11 11	11 11	11 12	11 11	11 11	11 11	11 11	11 11
D18S51	12 19	12 12	12 19	12 12	12 12	12 19	12 19	12 19	12 19
CSF1PO	11 12	11 11	12 12	11 12	11 12	11 12	11 12	11 11	11 12
D2S1338	18 19 20	19 20	19 20	18 19	20 20	19 20	19 20	19 20	19 20
Penta E	5 12	5 12	5 12	5 12	5 5	5 12	5 12	5 12	5 12
Penta D	8 11 12	8 11	11 11	8 11	8 11	11 12	11 12	11 12	8 11
SE33	19 25.2 28.2 29.2	19 25.2	19 25.2	28.2 29.2	25.2 28.2	25.2 29.2	19 25.2	28.2 29.2	25.2 29.2
D22S1045	11 15	11 11	11 15	11 15	11 15	11 15	11 15	11 15	11 11
D1S1656	12 16 17	12 17	12 17	16 17	12 17	16 17	12 16	12 16	12 17
D10S1248	12 14	12 12	12 14	14 14	12 14	14 14	12 14	12 14	12 12
D2S441	11 14	11 14	11 14	11 14	14 14	11 14	11 14	11 14	11 14
D12S391	19 21 22 23	19 19	19 22	22 23	19 22	21 22	19 21	19 21	19 22
LR		**1.3×10^{81}**	**9.9×10^{85}**	**1.8×10^{94}**	**4×10^{84}**	**2.3×10^{89}**	**2×10^{90}**	**9.5×10^{90}**	**2.3×10^{89}**

Figure 2: Profiles of all adventitious matches that could not be excluded from the mixed trace with 23-locus profile with their respective LR.

In an attempt to solve the case, profiling was performed with 28 STR systems by combining three multiplex STR kits: PowerPlex®16 HS, PowerPlex® ESI 17 and PowerPlex® CS7 (Promega Corporation; Madison, WI, USA).

By increasing the number of tested loci, three potential suspects were still considered (Figure 3). The statistical results confirmed the inclusion of the three suspects and none of the individuals could be excluded by any of the 28 tested loci.

Our efforts in the recent decade have been made to assess the degree of uncertainty in the analysis of STR profiles, in particular the mixed DNA profiles. In undertaking such a study, it is evidently necessary to have a known standard of true mixtures.

517 samples were collected randomly from Lebanese villages of different religious backgrounds. Profiling was performed with three different profile sizes: 16, 23 and 28 STR systems using three multiplex STR kits: PowerPlex® 16 HS, the PowerPlex® ESI 17 and the PowerPlex® CS7 (Promega Corporation; Madison, WI, USA). The obtained profiles were used to simulate DNA mixtures of two contributors. Each of the 517 profiles was then probed against the electronically simulated two-contributor mixtures (the same was done with each of the 16, 23 and 28 profile sizes). Mixture analysis was performed and Likelihood Ratios were generated whenever false inclusions were detected.

In the present case, profiles of individuals #3 and #6 were the known contributors of the mixture.

This given defies the statistical results generated by the DNA mixture analysis software that yielded LR values 1.3×10^{81}, 99×10^{85}, 4×10^{84}, 2.3×10^{89}, 9.5×10^{90} and 2.3×10^{89} for suspects 1, 2, 4, 5, 7 and 8 respectively with 23-locus profiles that supported their contribution and were sometimes higher than the LR values of the true contributors (Suspect 7, relative to suspect 6) [10,11].

Locus	Trace	Suspect 3	Suspect 6	Suspect 2
D3S1358	15 17 18	17 18	15 17	15 17
D19S433	13 16	13 16	13 16	13 16
D8S117	8 12 13 15	12 13	8 15	12 15
D5S818	11 13 14	13 14	11 11	11 13
TH01	7 9 10	7 10	9 10	9 10
vWA	14 20	14 20	20 20	14 20
D21S11	29 30 30.2	30 30.2	29 30	29 30
D13S317	10 12	10 12	12 12	12 12
TPOX	8 10 11	8 11	8 10	8 10
FGA	22 23	23 23	22 23	22 22
D7S820	9 10 12	9 10	10 12	10 10
D16S539	11 12	11 12	11 11	11 11
D18S51	12 19	12 12	12 19	12 19
CSF1PO	11 12	11 12	11 12	12 12
D2S1338	18 19 20	18 19	19 20	19 20
Penta E	5 12	5 12	5 12	5 12
Penta D	8 11 12	8 11	11 12	11 11
SE33	19 25.2 28.2 29.2	28.2 29.2	19 25.2	19 25.2
D22S1045	11 15	11 15	11 15	11 15
D1S1656	12 16 17	16 17	12 16	12 17
D10S1248	12 14	14 14	12 14	12 14
D2S441	11 14	11 14	11 14	11 14
D12S391	19 21 22 23	22 23	19 21	19 22
LPL	9 10	9 9	10 10	10 10
F13B	7 8 10	7 10	8 10	8 8
FESFPS	10 11 12	11 11	10 12	10 12
F13A01	5 6	5 5	6 6	5 6
Penta C	11 11	11 11	11 11	11 11
LR		**3×10^{97}**	**5×10^{92}**	**3×10^{96}**

Figure 3: Profiles of all adventitious matches that could not be excluded from the mixed trace with 28-locus profile with their respective LR.

Discussion and Conclusion

While increasing the number of STR loci up to 28, the power of discrimination increased and the possibility of false inclusions was reduced, but one individual remained falsely included in the DNA mixture of two other contributors. It is noteworthy that the LR was higher than the LR of one of these contributors. Even though relevant frequency datasets and an inbreeding coefficient were considered and expert DNA mixture analysis software was used, statistical analysis falsely supported the inclusion of non-contributors.

This case is likely to be encountered in a population with increased inbreeding practices. It is represented in order to highlight the risk of drawing conclusions in mixture analysis even if the relevant population and the inbreeding coefficient are accounted to in statistical analysis. The resulting false inclusions shed the light on the effect of allele sharing and homozygosity on the evaluation of DNA mixtures especially in consanguineous and endogamous populations. Presenting the DNA evidence without statistics renders the DNA evidence inadmissible. However, these findings challenge the admissibility of DNA mixture statistics, in particular in inbred communities and raise the attentiveness to forensic DNA mixture inclusive conclusions when dealing with communities with high-level of inbreeding.

Consequently, in similar populations, we recommend to restrain from establishing an inclusion interpretation in cases of mixed DNA traces, even when 28-locus profiles are used and statistical analysis is performed by expert software. However, such traces could be definitely used for exclusion purposes.

References

1. Torres Y, Flores I, Prieto V, Lopez-Soto M, Farfan MJ, et al. (2003) DNA mixtures in forensic casework: A 4-year retrospective study. Forensic Sci Int 134: 180–186.
2. Bieber FR, Buckleton JS, Budowle B, Butler JM, Coble MD (2016) Evaluation of forensic DNA mixture evidence: protocol for evaluation, interpretation, and statistical calculations using the combined probability of inclusion. BMC Genet 17: 125.
3. Gill P, Brenner CH, Buckleton JS, Carracedo A, Krawczak M, et al. (2006) DNA commission of the International Society of Forensic Genetics: Recommendations on the interpretation of mixtures. Forensic Sci Int 160: 90–101.
4. Ladd C, Lee HC, Yang N, Bieber FR (2001) Interpretation of complex forensic DNA mixtures. Croat Med J 42: 244–246.
5. Thompson WC, Mueller LD, Krane DE (2012) Forensic DNA statistics: Still controversial in some cases. The Champion 36: 12-23.
6. Al-Azem M, El-Andari A, Mansour I (2017) Estimation of allele and haplotype frequencies for 23 YSTR markers in the Lebanese population. Forensic Res Criminol Int J 5: 00150.
7. Abbas S, Mourad L, Mansour I (2018) Evaluation of sibling-ship analysis in secluded Lebanese villages with increased mating patterns. J Forensic Invest 6: 1-4.
8. El Andari A, Mansour I (2013) Is he or is he not the father? Pros, cons effect of following standardized interpretations. Forensic Sci Int: Genet Suppl Ser 4: e186–e187.
9. Evett IW, Weir BS (1998) Interpreting DNA evidence: Statistical genetics for forensic scientists. Sinauer Associates. Sunderland, Massachusetts, USA.
10. El-Andari A, Mourad L, Mansour I (2018) Genetic variations and population data on five supplementary STR markers in Lebanon. Ann Hum Genet 83: 1–4.
11. El-Andari A, Othman H, Taroni F, Mansour I (2013) Population genetics data from 23 STR markers from Lebanon. Forensic Sci Int Genet 7: e108–e113.