Rechtsmedizin 2025 · 35:22–28
https://doi.org/10.1007/s00194-024-00734-4
Accepted: 27 September 2024
Published online: 20 January 2025

DNA mixture deconvolution using fully continuous models EuroForMix and EFMrep

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Abstract

Background and objective: In the molecular genetic context, deconvolution describes the derivation of individual DNA profiles from a DNA mixture. Whereas mixture deconvolution is conventionally carried out manually by an expert, there are various probabilistic software applications to automate the process and two of these applications, EuroForMix (EFM) and EFMrep, are used in numerous forensic laboratories. They are particularly helpful when trace material is degraded or PCR artifacts are present and multiple replicates, if applicable from different DNA extracts, are combined for a better assessment. The aim of this study was to evaluate software-assisted deconvolution (EFM and EFMrep) with respect to correctly derived genotypes or alleles per profile in comparison to manual deconvolution based on a combination of at least two different DNA extracts per case.

Material and methods: To this end, 16 cases from former routine work were selected as examples and re-evaluated with EFM and EFMrep. In all cases, the manually derived profiles were incomplete due to the complexity of the underlying mixtures but still generated a hit in the German DNA Analysis Database (DAD). Likelihood calculations were performed for each match and the corresponding mixtures before the respective profiles were accepted as ground truth and the reference for deconvolution.

Results and conclusion: The results show that both forms of software are a useful addition to manual deconvolution, although they cannot replace it. Thus, when run in parallel they can objectively support the expert when there is uncertainty about an allele or genotype. In several cases it was shown that alleles were not correctly derived despite taking the threshold for the probability of results suggested in the current recommendations into account.

Keywords
Mixture deconvolution · Fully continuous software · EFMrep · EuroForMix · DNA mixture · Likelihood ratio

Supplementary Information

The online version of this article (https://doi.org/10.1007/s00194-024-00734-4) includes additional study data.

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Introduction

Mixture deconvolution is a powerful tool for inferring individual DNA profiles from DNA mixtures for subsequent transmission to a database or database queries. To carry out deconvolution, a mixed trace can be interpreted either manually by an expert or, for example, by a software based on a fully continuous (fc) model such as EuroForMix (EFM) or EFMrep [1–3]. Recommendations for the use of fc software for the biostatistical evaluation of forensic DNA analytical findings have already been published, e.g., by the German Stain Commission and the project group "Biostatistical DNA-calculations" in 2022. The recommendations stipulate a probability of at least 0.99, at or above which a derived genotype has the sufficient certainty to be reported [4, 5].

Mixed samples with degraded and low template DNA often produce electropherograms with dropouts or drop-ins and varying contributor proportions among loci. In such cases, multiple mix-

Table 1 Overview of the selected samples regarding the number of samples, number of replicates, the minimum number of contributors per sample and the number of known contributors

Case	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16
No. of samples	3	4	2	2	4	3	2	2	2	2	2	2	4	4	3	3
No. of replicates per sample	2	2	2	2	2	2	3	3	3	2	2	2	2	2	2	2
Minimum no. of contributors	3	3	4	3	3	3	3	3	3	3	3	3	2	3	3	2
No. of known contributors	0	1	1	1	1	1	0	0	0	0	0	1	0	1	1	1

ture samples stemming from the same trace (for example multiple vaginal swabs taken from one rape victim) or a trace complex can be taken into consideration for deconvolution to increase the number and certainty of the derived alleles. The EFM provides replicate-based deconvolution given that they were typed with the same multiplex and have identical mixture proportions, peak height distribution and degradation effects. With the extended model EFMrep it is now possible to specify the model parameters for individual samples and thus combine samples for analysis [1, 2]. Before EFMrep was released, EFM was used in our laboratory to corroborate tendencies the expert already had for the deconvolution of DNA mixtures based on more than one trace, although the software is not explicitly declared for this application. After the release of EFMrep, we re-evaluated the corresponding cases to see if and to what extent more alleles could be correctly deconvolved with sufficient certainty.

When deconvoluting a mixture, different experts should come to the same conclusion, which is why deconvolution results are always proofread by a second expert in our laboratory. Still, minor deviations can occur if, for example, one expert reports an allele that another expert is not certain about. The use of software such as EFM and EFMrep, on the other hand, should increase objectivity. The fc software assigns every allele in a mixture to different contributors with a certain probability based on imported data, considering peak heights, number of contributors, locus-specific amplification efficiency, an empirically determined, laboratory-specific drop-in probability as well as an estimated dropout probability, degradation effects and stutter events [6].

The aim of this study was to evaluate whether software-aided deconvolution produces more complete and correct profiles in comparison with manual deconvolution based on the recommendations issued by the German Stain Commission and the project group "Biostatistical DNA-calculations" mentioned above [5].

Material and methods

Case selection

For this study 16 cases were re-evaluated in which DNA mixtures were deconvolved based on multiple extracts and their corresponding replicates. The traces were considered suitable for combination because they were each taken from coherent evidence, for example multiple vaginal swabs taken from one woman or DNA stubs used on one piece of clothing, where the same mixture contributors could be consistently detected. None of the deconvolved alleles were present in a ratio of 4:1 (or larger) compared to the remaining mixture contributors and therefore could not be classified as major contributors in the corresponding electropherograms [7]; however, in most cases the derived alleles were either more prominent than others or the mixtures included known contributors (for example the victim), which is why the expert was able to perform deconvolution. In all cases, at least one allele could not be derived with sufficient certainty; however, each profile matched with an individual dataset stored in the DAD. Additionally, the likelihood ratio for the opposing hypotheses "the derived profile contributed to the mixture" and "the derived profile did not contribute to the mixture" was calcula–9ted for each case. All results were above 3.5 E + 11 and thus accepted as the ground truth for each case [8].

Table 1 shows an overview of the cases based on the number of samples taken from the same trace or trace complex, the number of replicates per sample as well as the minimum number of contributors and the number of known contributors. Samples from cases 7–9 were particularly challenging and therefore genotyped three times instead of just twice. The minimum number of contributors was originally determined manually by the expert based on composite profiles of each sample used for deconvolution [9]. All values were confirmed by the software using the "automatic model search" option, which calculates the combination of number of contributors, degradation and stutter effects that best explains the respective electropherogram.

Genotyping

DNA extraction of all samples was performed using the Maxwell® RSC Blood DNA kit or Maxwell® FSC DNA IQ™ Casework kit and the Maxwell® RSC Instrument (Promega, Madison, WI, USA). DNA was quantified with the Quantifiler™ Trio DNA Quantification kit on a 7500 Real Time PCR System (Thermo Fisher Scientific, Waltham, MA, USA) and subsequently amplified on various Thermal Cyclers (Thermo Fisher Scientific) using PowerPlex® ESX 17 fast System (Promega). Capillary electrophoresis was performed on a 3500 × L Genetic Analyser (Thermo Fisher Scientific). All samples were analyzed using the GeneMapper™ ID-X software (up to version v 1.4). A detection threshold of 100 rfu was applied for all dye channels. Marker-specific stutter ratios were applied as specified by the manufacturer.

Deconvolution

Deconvolution was originally carried out manually by an expert and reviewed by a second. Wherever the expert was not sufficiently certain about a single allele or a genotype it was not reported. This is comparable to a software result with a probability of less than 0.99 [4, 5]. The

Table 2 Overview of genotypes derived manually and using EuroForMix and EFMrep

Expert		EFM				EFMrep			
Complete and correct GT	Not specified	Correct GT		False GT		Correct GT		False GT	
194	62	241		15		246		10	
–		$p \geq 0.99$	$p < 0.99$	$p \geq 0.99$	$p < 0.99$	$p \geq 0.99$	$p < 0.99$	$p \geq 0.99$	$p < 0.99$
		186	55	3	12	211	35	3	7

minimum number of contributors per sample was determined using the maximum likelihood function provided by the software.

The EuroForMix version 4.0.8 and EFMrep version 1.0.1 were used. Both EFM and EFMrep were set to a probability of drop-in of 0.0056 and a drop-in hyperparameter of 0.025 according to in-house validation [10]. The backward and forward stutter option was deactivated, and the analytical threshold was not specified as data were preprocessed regarding both parameters due to GeneMapper settings. Deconvolution with each software was performed based on European frequencies provided by the "STRs for Identity ENFSI Reference" (STRidER 2.0) database, hosted by the European Network of Forensic Science Institutes (ENFSI). For both EFM and EFMrep, "ESX17fast" was selected for all samples according to prior laboratory work.

The results were assessed in terms of complete genotypes per locus (two alleles for both heterozygous and homozygous genotypes (GT)) and alleles per locus. Comparison of the results obtained by the expert and using both EFM and EFMrep was based on the following criteria: the number of derived genotypes and alleles per locus with the recommended probability of 0.99 or greater as well as correctness, regarding the ground truth of each derivation.

Results and discussion

Deconvolved genotypes

Altogether, 16 cases and 16 loci per case resulted in the derivation of 256 genotypes.

Table 2 shows the total number of correct and false genotypes interpreted with EFM and EFMrep. Correct genotypes derived via software are subdivided into probabilities at least equal to or below 0.99, which is the recommended threshold for valid predictions. The expert did not derive any wrong GT but decided to report 1 allele at most 62 times.

The results show that EFM deduced 241 correct GT and EFMrep 246. The expert reported 194 complete and correct GT; however, these values include GT that although predicted correctly, did not reach the recommended threshold. Hence, for performance comparison, only the software results with p greater than or equal to 0.99 can be compared with the GT derived by the expert.

However, considering the number of correct genotypes that were predicted with a probability of at least 0.99, EFM achieves 8 results less and EFMrep 17 results more than the expert. The probability of the results is increased for EFMrep due to fact that EFMrep, in comparison to EFM, distinguishes between imported individual samples and their assigned replicates. Therefore, two samples can differ greatly from each other in terms of mixture proportions, peak height imbalances and drop-in or drop-out events, despite stemming from the same trace complex or even the same trace [11]. The EFMrep algorithm takes this into account based on the settings provided by the user and consequently reacts more robustly to deviations that are not to be expected among replicates. The EFM and EFMrep differed only slightly regarding the number of falsely deduced GT altogether (15 for EFM, 10 for EFMrep) and both software falsely deduced 3 GT with a probability of 0.99 or greater. This indicates that particularly challenging samples may be difficult to assess with either software.

Figure 1 (**Fig. 1**) exemplarily shows a summary of the genotypes derived manually and by both EFM and EFMrep along with the corresponding probabilities (rounded to two decimal places) for two cases. Only genotypes that were correctly predicted (green) with a probability below 0.99 (light green) as well as genotypes falsely predicted ($p \geq 0.99$ dark red and $p < 0.99$ light red) are shown for a better overview. Additionally, genotypes that were not or not completely derived by the expert are marked in blue. One "?" represents a missing allele, "?/?" resembles a missing genotype (heterozygous and homozygous, respectively). The corresponding DAD match and, in accordance with LR calculations, assumed ground truth is highlighted in grey.

Fig. 1 ▶ GT-table for exemplarily selected cases 1 and 5. DAD-match and "ground truth" are shown in the first line of each case in *grey*. The second line includes the results of manual deconvolution, *blue* highlights GT the expert did not completely report. GT (EFM) and GT (EFMrep) show deconvolution results of the respective software, correct GT and false GT are highlighted in *green* and *red*, respectively. All correctly derived GT are highlighted in light ($p < 0.99$) and dark ($p \geq 0.99$) green, respectively. All incorrectly derived GT are highlighted in *light* ($p < 0.99$) and *dark* ($p \geq 0.99$) *red*, respectively. Empty cells represent correct deductions with $p \geq 0.99$ (data not shown for better overview of the table). GT derived correctly by the expert are not highlighted.

Deconvolution in case 1 was based on three different extracts with two replicates each. The minimum number of contributors was set to three, all of which were unknown prior to analysis. Whereas the expert reported only one allele in FIBRA due to very small peak heights for allele 25, the complete GT was deduced correctly with both software with $p \geq 0.99$. The GT for D21S11 (EFM) and D2S441 (EFM and EFMrep) were incorrectly interpreted as heterozygous by EFM, although both GT were in fact homozygous according to the respective DAD match. Notably, both incorrectly derived GT were originally reported correctly by the expert. The misinterpretations in D21S11 can be explained with varying mixture proportions between the included samples, as **Fig. 2** exemplarily shows. This is consistent with the fact that EFM was not designed to process samples from multiple extracts.

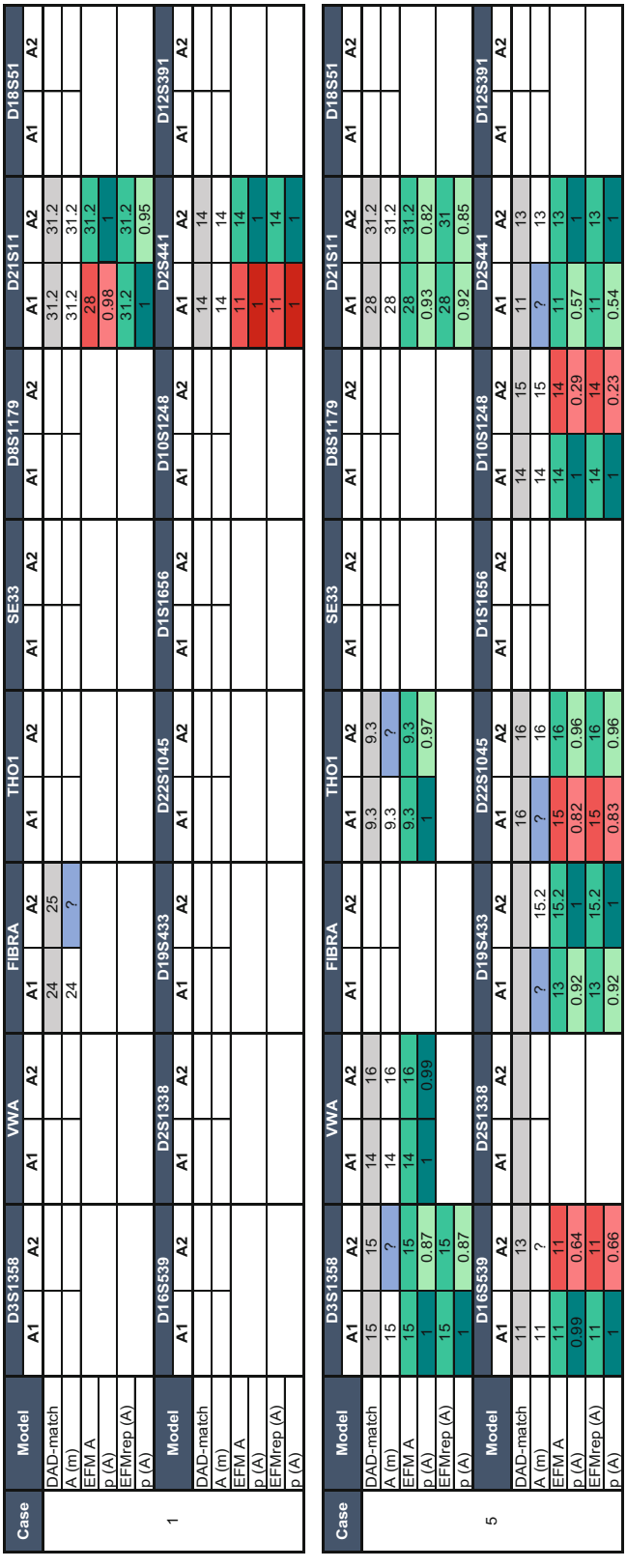
The incorrect deduction for D2S441, on the other hand, may have been caused by the necessity to assign allele 11 to a contributor to maintain the number of contributors of three; however, the fact that the

EFM and EFMrep Genotypes and Probabilities

Case	Model	D3S1358	VWA	FIBRA	THO1	SE33	D8S1179	D21S11	D18S51	D16S539	D2S1338	D19S433	D22S1045	D1S1656	D10S1248	D2S441	D12S391
1	DAD-match			24 / 25				31.2 / 31.2								14 / 14	
	GT (m)			24 / ?				31.2 / 31.2								14 / 14	
	GT (EFM)							28 / 31.2								11 / 14	
	p (GT)							0.98								1	
	GT (EFMrep)							31.2 / 31.2								11 / 14	
	p (GT)							0.95								1	
5	DAD-match	15 / 15	14 / 16		9.3 / 9.3			28 / 31.2		11 / 13		13 / 15.2	16 / 16		14 / 15	11 / 13	
	GT (m)	15 / ?	14 / 16		9.3 / ?			28 / 31.2		11 / ?		15.2 / ?	16 / ?		14 / 15	13 / ?	
	GT (EFM)	15 / 15	14 / 16		9.3 / 9.3			28 / 31.2		11 / 11		13 / 15.2	15 / 16		14 / 14	11 / 13	
	p (GT)	0.86	0.96		0.97			0.75		0.63		0.92	0.78		0.65	0.56	
	GT (EFMrep)	15 / 15						28 / 31.2		11 / 11		13 / 15.2	15 / 16		14 / 14	11 / 13	
	p (GT)	0.87						0.77		0.66		0.92	0.79		0.74	0.54	

Fig. 2 ▲ Electropherogram excerpts of locus D21S11 (case 1) exemplarily show varying mixture proportions that may have led to the incorrect assignment of the genotype 28/31.2

Fig. 3 ▲ Electropherogram excerpts of locus D10S1248 (case 5) exemplarily show peak height imbalances that may have led to the incorrect assignment of the genotype 14/14



Case	Model	D3S1358		VWA		FIBRA		THO1		SE33		D8S1179		D21S11		D18S51	
		A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2
1	DAD-match					24	25							31.2	31.2		
	A (m)					24	?							31.2	31.2		
	EFM A													28	31.2		
	p (A)													0.98	1		
	EFMrep (A)													31.2	31.2		
	p (A)													1	0.95		
	Model	D16S539		D2S1338		D19S433		D22S1045		D1S1656		D10S1248		D2S441		D12S391	
		A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2
	DAD-match													14	14		
	A (m)													14	14		
	EFM A													11	14		
	p (A)													1	1		
	EFMrep (A)													11	14		
	p (A)													1	1		

Case	Model	D3S1358		VWA		FIBRA		THO1		SE33		D8S1179		D21S11		D18S51	
		A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2
5	DAD-match	15	15	14	16			9.3	9.3					28	31.2		
	A (m)	15	?	14	16			9.3	?					28	31.2		
	EFM A	15	15	14	16			9.3	9.3					28	31.2		
	p (A)	1	0.87	1	0.99			1	0.97					0.93	0.82		
	EFMrep (A)	15	15											28	31		
	p (A)	1	0.87											0.92	0.85		
	Model	D16S539		D2S1338		D19S433		D22S1045		D1S1656		D10S1248		D2S441		D12S391	
		A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2	A1	A2
	DAD-match	11	13					16	16			14	15	11	13		
	A (m)	11	?			?	15.2	?	16			14	15	?	13		
	EFM A	11	11			13	15.2	15	16			14	14	11	13		
	p (A)	0.99	0.64			0.92	1	0.82	0.96			1	0.29	0.57	1		
	EFMrep (A)	11	11			13	15.2	15	16			14	14	11	13		
	p (A)	1	0.66			0.92	1	0.83	0.96			1	0.23	0.54	1		

Fig. 4 ◀ Allele table for exemplarily selected cases 1 and 5. The color scheme is identical to the one in **Fig. 1**. Additionally, correct alleles with $p \geq 0.99$ are highlighted in *dark green*

software interpreted the corresponding GT with a probability of $p = 1$ is misleading and could result in an incorrect genotype in a database report at worst. This highlights that checking the results for plausibility is essential. In case 5, duplicates of four different samples with three mixture contributors were used for deconvolution, whereby the DNA profile of one person was known and imported into the software in advance. The fact that the expert reported only one allele in six loci indicates that the mixtures were difficult to assess, which is reflected in the fact that neither of the software could correctly interpret the GT marked in blue with sufficient certainty; however, the four different mixtures were very similar in terms of contributor proportions, which explains the comparable results between both software despite the high number of insufficient certainty ($p < 0.99$) and errors. The incorrect results in systems D16S539, D22S1045 and D10S1248 given by both software were presumably caused by peak height imbalances, which also led the expert to report only one of two alleles for D16S539 and D22S1045 each. **Figure 3** shows two electropherogram excerpts of locus D10S1248 for visualization.

Furthermore, as shown in **Fig. 1**, all GTs marked in red were derived as either false homozygous or false heterozygous, although the true GT is opposite in each case. To investigate whether this type of misinterpretation occurred more frequently than other types of errors, a one-sided independent t-test was performed including all samples ($n = 16$) but the test result was not significant with $t(18) = 0.23$, $p = 0.41$.

Altogether, software-aided interpretation of the mixed traces enabled the correct ($p > 0.99$) derivation of GT that were not previously reported by the expert in 21 (EFM) and 27 (EFMrep) systems, respectively, spread over 11 cases (data shown in the Supplementary Information online). The EFM deduced wrong GT that were not or not fully derived by the expert eight times, and EFMrep seven times. Furthermore, the occurrence of three (EFM) or four (EFMrep) incorrect but sufficiently certain derivations according to the recommendations is an indication that the software's

Table 3 Overview of GT derived manually and using EFM and EFMrep

Expert		EFM				EFMrep			
Correct A	Not specified	Correct A		False A		Correct A		False A	
459	53	497		15		502		10	
–		$p \geq 0.99$	$p < 0.99$	$p \geq 0.99$	$p < 0.99$	$p \geq 0.99$	$p < 0.99$	$p \geq 0.99$	$p < 0.99$
		439	58	4	11	465	37	3	7

findings should always be assessed critically.

Additionally, we assessed the *p*-values for interpreted results given by EFM and EFMrep. All correct genotypes with $p < 0.99$ were subsequently evaluated in more detail to find out whether at least one allele could be determined correctly and with a minimum probability of 0.99.

Deconvolved alleles

In analogy with **Table 2**, **Table 3** presents the overall results in terms of derived alleles. Altogether, the expert reported 459 correct alleles.

Deconvolution with EFM and EFMrep, on the other hand, derived 497 and 502 correct alleles, respectively. Again, the number of correctly derived alleles does not equal the number of alleles suitable for reporting. A total of 88.3% (EFM) or 92.6% (EFMrep) of the correctly predicted alleles met or exceeded the recommended threshold for *p* of 0.99 which equals 20 less and 6 in addition, respectively, compared to the ones derived by the expert.

Figure 4, in analogy to **Fig. 1**, shows a summary of the alleles derived by the expert and EFM as well as EFMrep along with the corresponding probabilities (rounded to two decimal numbers) for cases 1 and 5. Blank spaces resemble alleles which were predicted correctly.

In cases 1 and 5 (**Fig. 4**) as well as all other evaluated cases, one correct allele could be deduced for an initially incorrect GT. Furthermore, in 59 out of 63 loci (EFM) and 36 out of 40 loci (EFMrep), 1 allele per STR system could be derived with a probability equal to or greater than 0.99. With overall five exceptions, all the alleles that were not conclusively derived by the expert, coincided with those that were determined correctly but not with a sufficient degree of probability by the software. These alleles remain uncertain due to constellations of intersample as well as intrasample and intralocus peak height imbalances and shared alleles between individual contributors.

Conclusion

Neither of the software we tested can replace the expert but both EFM and EFMrep can support manual deconvolution of complex mixed traces. Prior to deconvolution via software, electropherograms should be carefully reviewed with respect to their suitability for software-supported assessment.

We found the recommended *p*-value of 0.99 to be an overall reasonable benchmark for the results of mixture deconvolution with EFM and EFMrep; however, in some cases it was shown that despite applying the threshold, alleles were not correctly derived. Hence, thorough review of the results is mandatory.

The software should be used after manual deconvolution and based on the prediction of single alleles instead of genotypes per locus. Thus, prevailing expert's tendencies can be supported objectively.

Although it was not designed for this purpose, EFM gives reliable results when the combined samples show good comparability of mixture ratios and degradation effects and takes significantly less time for the calculation process.

Our results show that EFMrep is a very useful extension of the EFM software in cases where the deconvolution of an individual profile is performed in consideration of replicates of different DNA extracts.

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Author Contribution. All authors contributed to the study conception and design. Material preparation, data collection and analysis were performed by R. Schiller, J. Fleckhaus, D. von Máriássy, V. Brune, B. Bayer and K. Anslinger. The first draft was written by R. Schiller and all authors commented on subsequent versions of the manuscript. All authors read and approved the final manuscript.

Funding. Open Access funding enabled and organized by Projekt DEAL.

Data availability. All data supporting the findings of this study are available within the paper and its supplementary information.

Declarations

Conflict of interest. R. Schiller, J. Fleckhaus, D. von Máriássy, V. Brune, B. Bayer and K. Anslinger declare that they have no competing interests.

The authors did not conduct any studies on humans or animals for this article.

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Zusammenfassung

Ableitung von DNA-Identifizierungsmustern aus DNA-Mischungen unter Verwendung der vollkontinuierlichen Modelle EuroForMix und EFMrep

Hintergrund und Zielsetzung: Im molekulargenetischen Kontext beschreibt Dekonvolution die Ableitung einzelner DNA-Profile aus einer DNA-Mischspur. Während die Ableitung konventionell manuell von einem Sachverständigen durchgeführt wird, gibt es verschiedene probabilistische Softwareanwendungen, um den Prozess zu automatisieren. EuroForMix (EFM) und EFMrep werden als zwei dieser Anwendungen in zahlreichen forensischen Laboren genutzt. Sie sind besonders dann hilfreich, wenn Spurenmaterial degradiert ist oder PCR-Artefakte vorliegen und mehrere Replikate, ggf. auch aus mehreren DNA-Extrakten, zur besseren Beurteilung kombiniert werden. Ziel dieser Studie war die Evaluation softwaregestützter Ableitung (EFM und EFMrep) hinsichtlich korrekt abgeleiteter Genotypen bzw. Allele pro Profil im Vergleich zur manuellen Ableitung auf Grundlage einer Kombination aus mindestens zwei verschiedenen DNA-Extrakten pro Fall.
Material und Methoden: Zu diesem Zweck wurden 16 Fälle aus der ehemaligen Routinearbeit als Beispiele ausgewählt und mit EFM und EFMrep neu ausgewertet. In allen Fällen waren die manuell abgeleiteten Profile aufgrund der Komplexität der zugrundeliegenden Mischungen unvollständig, erzeugten aber dennoch einen Treffer in der deutschen DNA-Analyse-Datenbank (DAD). Für jedes getroffene Profil und die entsprechenden Mischungen wurden Likelihood-Berechnungen durchgeführt, bevor die jeweiligen Profile als Grundwahrheit und Referenz für die Ableitung akzeptiert wurden.
Ergebnisse und Schlussfolgerung: Die Ergebnisse zeigen, dass beide Softwares eine sinnvolle Ergänzung zur manuellen Ableitung darstellen, auch wenn sie diese nicht ersetzen können. Somit können sie parallel ausgeführt den Sachverständigen objektiv unterstützen, wenn dieser bezüglich eines Allels oder Genotyps unsicher ist. In mehreren Fällen konnte gezeigt werden, dass trotz Berücksichtigung des in den aktuellen Empfehlungen vorgeschlagenen Schwellenwerts für die Wahrscheinlichkeit abgeleiteter Merkmale Allele nicht korrekt abgeleitet wurden.

Schlüsselwörter
Dekonvolution von Mischspuren · Vollkontinuierliche Software · EFMrep · EuroForMix · DNA-Mischung · Likelihood Ratio