



GHEP-ISFG Proficiency Test 2011: Paper challenge on evaluation of mitochondrial DNA results

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ABSTRACT

In this GHEP-ISFG exercise, participating labs were invited to evaluate a forensic case in which the mtDNA haplotype from a hair shaft in the victim's hand matched the suspect's haplotype. 31 forensic labs participated in the exercise. Although all except one used the EMPOP database to estimate the haplotype frequencies different final likelihood ratios (LRs) were reported. The main factors affecting these differences were: the origin of the reference population, the approaches to correct sampling errors, the LR formula, the source of EMPOP data (forensic/literature), the type of search (pattern or literal and "disregard Indels" option) and the selected edition range to perform the queries. This demonstrates that further efforts are needed in order to standardize the statistical evaluation of the mtDNA evidence.

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1. Introduction

Likelihood ratio (LR), based on the frequency that specific genetic data appear in a reference population is the recommended way to evaluate DNA analysis results in Europe. In the case of mtDNA, some of the parameters which most influence the estimates of haplotype frequencies are: (i) edition range of the query, (ii) inclusion/exclusion of point and/or length heteroplasmies in the searches, (iii) selection of the reference population and (iv) methodology to correct sampling errors in the database.

The Spanish and Portuguese Speaking Group of the International Society for Forensic Genetics (GHEP-ISFG) has shown to have a big interest in the use and standardization of the mtDNA analysis with forensic purposes ([1–4], among others). In order to obtain a realistic picture of the communication of mtDNA evidence at court, the GHEP-ISFG has proposed an exercise regarding the interpretation of mtDNA results in its last Proficiency Test 2011.

2. Materials and methods

Laboratories were asked to evaluate a common simulated forensic case (Table 1). The edition range included the whole Control Region (16024–576). Labs were also asked to calculate the likelihood ratio (LR) under the following two hypotheses:

- (a) *H_p*: the hair shaft comes from the suspect or from a maternal relative of the suspect.
- (b) *H_d*: the hair shaft comes from a random individual of the European population not related to the suspect.

Two additional questions were put to the labs in order to better evaluate their results: (i) what mtDNA database they have used and (ii) the number of matches that were found in that database (e.g.: 7 matches of 4000 haplotypes).

3. Results

In total, 31 labs participated in this GHEP-ISFG exercise. All labs except one performed their searches in the EMPOP database (version 2.1, Release 4) [5]. Regarding the final figure after

Table 1

Wording of the proposed exercise: in a place located in Barcelona a homicide was committed. A hair shaft was recovered from one hand of the victim. Reference samples from the victim and suspect are available. After analyzing the entire mtDNA Control Region (16024–576) the results are shown in this table.

Sample	Edition range	Haplotype
Victim	16024–576	263G 315.1C 16519C
Suspect	16024–576	72C 263G 315.1C 16298C
Hair shaft	16024–576	72C 263G 315.1C 16298C

calculating the LR, three main groups can be distinguished. The first group (with LR around 200–241) considered the whole database (all populations); the second one (with LRs around 94–124) considered European population and the last group (LR = 32) considered only the Spanish population as reference. Three other labs reported different results out of these ranges.

4. Discussion

As far as we know, this is the first collaborative exercise regarding the statistical evaluation of the mtDNA evidence. We have selected a very easy simulated forensic case in order to know how forensic labs deal with this matter. The EMPOP database was the most used to evaluate mtDNA results, but the results showed some dispersion mainly due to the following causes (see Fig. 1):

4.1. The reference population

A high number of labs compared the mitochondrial haplotype with the entire world population available on database although in the wording of the hypothesis of the defense (*H_d*) in this exercise, labs were asked to evaluate the results in the European population.

4.2. Correction methods

All the resulting LRs except one were inside the expected range (LR = 200–241 for the labs using all EMPOP populations; and LR = 94–124 for labs using the EMPOP European population). In this exercise the fact of using one or other correction method has not had a big influence in the final result (except for one lab) but

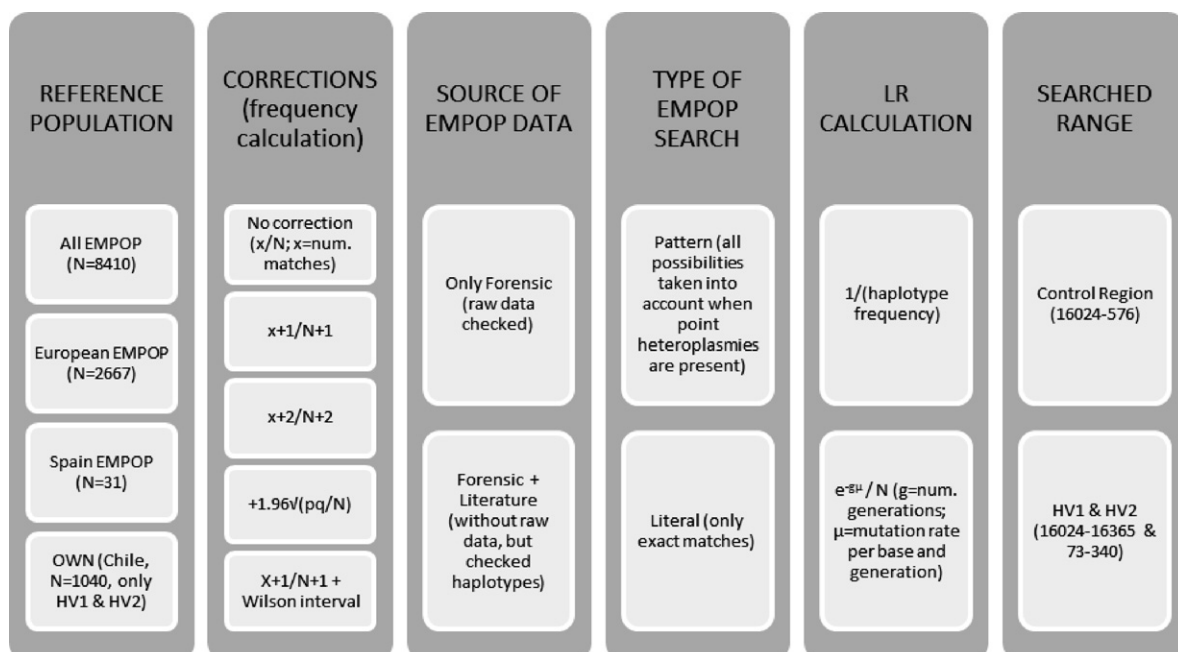


Fig. 1. Most influencing parameters in the final LR results of participating labs.

the influence that each method can have is very different depending on the number of coincidences in the database [6].

4.3. Source of data and type of queries

We would like to emphasize that “literature data” is as reliable as the “forensic” one since they have been submitted to many filters in order to detect possible errors. Also pattern searches are always preferred when evidence has to be statistically evaluated since they are more conservative than the “literal” searches. In this exercise the influence of using either “pattern” or “literal” queries was not very important due to the involved haplotype. Two labs performed the search without ignoring length variants in the HV2 homopolymeric tract and reported lower estimates of the haplotype frequency and therefore higher LR.

4.4. LR calculation

As no differences between evidence and suspect haplotypes were present, the LR ($p(E/Hp)/p(E/Hd)$) could be directly calculated as $1/(\text{haplotype frequency})$. One lab calculated the LR by using the formula $e^{-gH}/(\text{haplotype frequency})$ but only slight differences were shown when comparing with other labs. Perhaps, we should estimate the mutation rate not only regarding the number of generations or years, but also in each type of tissue/fluid to better evaluate the results in both kinship and criminal cases.

5. Conclusions

With the present exercise, we have managed to take a step forward in the estimates of haplotype frequencies from mtDNA databases and in the statistical interpretation of mtDNA results.

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Conflict of interest

None.

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