Available online at www.sciencedirect.com
ScienceDirect
Forensic Science International: Genetics Supplement Series 1 (2008) 654–655
www.elsevier.com/locate/FSIGSS

Research article

Effects of reference population and number of STR markers on paternity testing

Rossana Moroni [a,b,*], Dario Gasbarra [b], Elja Arjas [a,b], Matti Lukka [a], Ismo Ulmanen [a]

[a] *Paternity Testing Laboratory, National Public Health Institute, Helsinki, Finland*
[b] *Department of Mathematics and Statistics, University of Helsinki, P.O.Box 68, Gustaf Hällströmin katu 2b, Helsinki, FIN-00014, Finland*

Received 21 August 2007; accepted 10 October 2007

Abstract

Three sets of commonly used autosomal short tandem repeat (STR) markers (containing 15, 10 and 9 markers) and 14 databases from populations belonging to Africa, America, Asia and Europe were used to investigate how the selection of the population database and the number of considered markers would influence the statistical evidence that is usually produced to favour paternity. The study was based on a sample of 100 randomly chosen Finnish paternity trios collected during paternity testing case work and without any exclusion after use of 15 STR markers. Paternity index, power of evidence, typical paternity index and Random man excluded were computed and descriptive statistics were provided separately for trios (mother, child, alleged father) and duos (obtained from trios but not considering the genetic information of the mother). This was done for all combinations of markers and databases. In trio cases the differences between results obtained are not statistically significant. However, especially in duo cases the use of 15 STR markers is recommended.

Keywords: Paternity testing; Reference population; Database effect; Marker effect

1. Introduction

In recent years a growing number of disputed paternities have involved parents from different ethnic backgrounds. We examined how sensitive the paternity index (PI) [1] the power of evidence (PE) [1] and the random man excluded (RME) [1] are to the selection of the population STR database ('database effect') and to the number of considered STR markers ('marker effect'). To evaluate this we performed paternity tests, on 100 Finnish trios and duos with three sets of commonly used STR markers (containing 15, 10 and 9 markers) and 14 population databases, representing different marker allele frequencies. Concerning the 'database effect' we carried out a comparative statistical analysis of PI and PE, which were at first calculated considering the allele frequencies of the alleged father's own population, and then using the other reference population databases. A good measure of this effect is obtained from the analysis of the typical paternity index (TPI) [2]. Concerning the 'marker effect' we investigated the changes in the PI when performing the test using only the Finnish allele frequencies but considering three different sets of markers.

2. Materials and methods

A total of 100 Finnish standard 'Trios' paternity cases with no genetic inconsistencies between mother and child or between father and child were collected during paternity testing case work. The genotyping of the STR-loci was performed with AmpF*l*STR Profiler kit (9 markers), AmpF*l*STR SGM Plus kit (10 markers) and with combination of Profiler and SGM kits (15 markers). Database of allele frequencies for 14 populations (Finland, Poland, Turkey, Vojvoidina, Extremadura, Italy, Belgium, Kosovo, Mexico, Taiwan, Korea, El Salvador, Somalia, Mozambique) was collected from studies that have been already published. Parameters for paternity were computed, in every trio and in every duo (motherless cases, obtained from trios but not considering the genetic information of the mother) for all combinations of markers and databases. In details Paternity index (PI), Power of evidence (PE), typical paternity index (TPI) and random man excluded (RME) were computed and descriptive statistics performed. In order to have a measure of

* Corresponding author. Tel.: +358 9 191 51411; fax: +358 9 191 51400.
E-mail address: rossana.moroni@helsinki.fi (R. Moroni).

1875-1768/$ – see front matter
doi:10.1016/j.fsigss.2007.10.102

the database effect we compared TPI of different populations with Finnish TPI. TPI is the harmonic mean of PIs and it is considered to be a good measure of typical performance. Nei's distance was calculated between the Finnish population and all the other 13 populations to investigate if the PI is influenced by the genetic distance between reference population and the putative father's population.

3. Results and conclusions

3.1. Database effect

Trio cases. The results suggest that for trio cases there is no urgent need of using a specific population database, no matter what set of markers is in use, as the power of evidence is always greater than 0.99, 0.999 and 0.9999 for 9, 10 and 15 markers, respectively. The power of evidence and the random man excluded display similar distribution.

Duo cases present a notable database effect and values of power of evidence and random man excluded being significantly smaller than what we observed in the trio cases. Using 15 markers the PE is always greater than 0.99 but considering 10 and 9 markers (and all the different populations) we had 27 and 615 cases, respectively with PE smaller than 0.99. In details, of those cases, 1 and 183, respectively, present PE smaller than 0.95. It is recommendable to use a kit with the highest number of markers, in order to achieve reliable results. Interestingly, in some individual duo cases with 9 or 10 markers the PE and RME values displayed rather prominent differences.

3.2. Marker effect

Trio cases are weakly influenced by the number of considered markers. In Finnish population in duo cases, using 10 and 9 markers, 6 and 73 cases, respectively yield the power of evidence smaller than 0.99, which is the commonly used threshold value. In details, using the 9 markers set, 20 duo cases present a power of evidence smaller than 0.95. Results suggest that dealing with a reduced amount of information, typical of duo cases, it is advisable to use a 15 markers set.

Conflict of interest

None.

References

[1] I.W. Evett, B.S. Weir, Interpreting DNA evidence, Statistical Genetics for Forensic Scientists, Sinauer, 1998.

[2] C. Brenner, J.W. Morris, Paternity Index calculations in single locus hypervariable DNA probes: validation and other studies, in: Proceedings for the International Symposium on Human Identification, Promega Corporation, 1989.