Forensic Science International: Genetics Supplement Series 3 (2011) e238–e239

Contents lists available at ScienceDirect

Forensic Science International: Genetics Supplement Series

journal homepage: www.elsevier.com/locate/FSIGSS

Italian population data for the new ENFSI/EDNAP loci D1S1656, D2S441, D10S1248, D12S391, D22S1045. The GeFI collaborative exercise and concordance study

C. Previderè [a,*], P. Grignani [a], S. Presciuttini [b]

the GeFI Group

[a] Department of Legal Medicine, Forensic and Pharmaco-Toxicological Sciences, University of Pavia, Italy
[b] Department of Physiological Sciences, University of Pisa, Italy

ARTICLE INFO

Article history:
Received 29 August 2011
Accepted 31 August 2011

Keywords:
miniSTR
Database
Population study

ABSTRACT

A collaborative exercise of the ISFG Italian Working Group GeFI was organised to investigate the five new ENFSI/EDNAP miniSTR loci D1S1656, D2S441, D10S1248, D12S391 and D22S1045. Allele frequencies were determined in a sample of 960 individuals collected by the nineteen participating laboratories. The concordance of the genotypes has been evaluated in duplicate experiments, using different kits. No discordant genotypes were revealed for the five ENFSI/EDNAP miniSTR markers. All the labs participating in the collaborative exercise correctly typed the two blind blood stains sent for proficiency testing.

1. Introduction

Five new ENFSI/EDNAP loci, D1S1656, D2S441, D10S1248, D12S391 and D22S1045, were investigated in Italy in a collaborative exercise organized by the ISFG Italian Working Group GeFI. The inclusion of these five miniSTR markers in the European Standard Set of loci (ESS) was strongly recommended by ENFSI/EDNAP in order to avoid the possibility of random matches in international database exchanges. In addition, because of their shorter PCR products, the miniSTR markers have been shown to increase the number of degraded or low template samples which could be successfully investigated. However, population data for these new markers are limited. This collaborative exercise was organized in order to provide a large Italian database for the new miniSTR markers useful in forensic case analysis. The concordance of the genotypes will be evaluated in duplicate experiments, using different kits. A questionnaire aimed at investigating the analytical procedures has been sent to each participating laboratory.

2. Materials and methods

2.1. Samples

Nineteen laboratories (18 from universities and hospitals and 1 from a national criminal justice service) participated in the collaborative exercise, each one typing at least 50 unrelated subjects living in their regions, for a total of 960 samples. Saliva or blood was collected.

2.2. DNA extraction

Different DNA extraction methods were indicated by the labs. Qiagen procedure was the most used followed by phenol/chloroform, Chelex and magnetic beads. Other commercial kits were also indicated.

2.3. DNA quantification

Fourteen labs quantified the DNA extracts before STR analysis using UV spectrophotometry or qPCR.

2.4. Concordance study and STR analysis

Each laboratory was requested to type its samples in duplicate, freely choosing a combination of at least two of the following kits: PowerPlex ESX, PowerPlex ESI (Promega), AmpFlSTR NGM (Applied Biosystems), and Investigator ESSplex (Qiagen).

2.5. DNA electrophoresis

The amplified products were run on ABI 310, 3130 and 3500 Genetic Analysers (Applied Biosystems). Most of the labs analyzed the electropherograms using the GeneMapper software (Applied

* Corresponding author. Tel.: +39 0382 987820; fax: +39 0382 528025.
E-mail address: previde@unipv.it (C. Previderè).

1875-1768/$ – see front matter © 2011 Elsevier Ireland Ltd. All rights reserved.
doi:10.1016/j.fsigss.2011.08.118

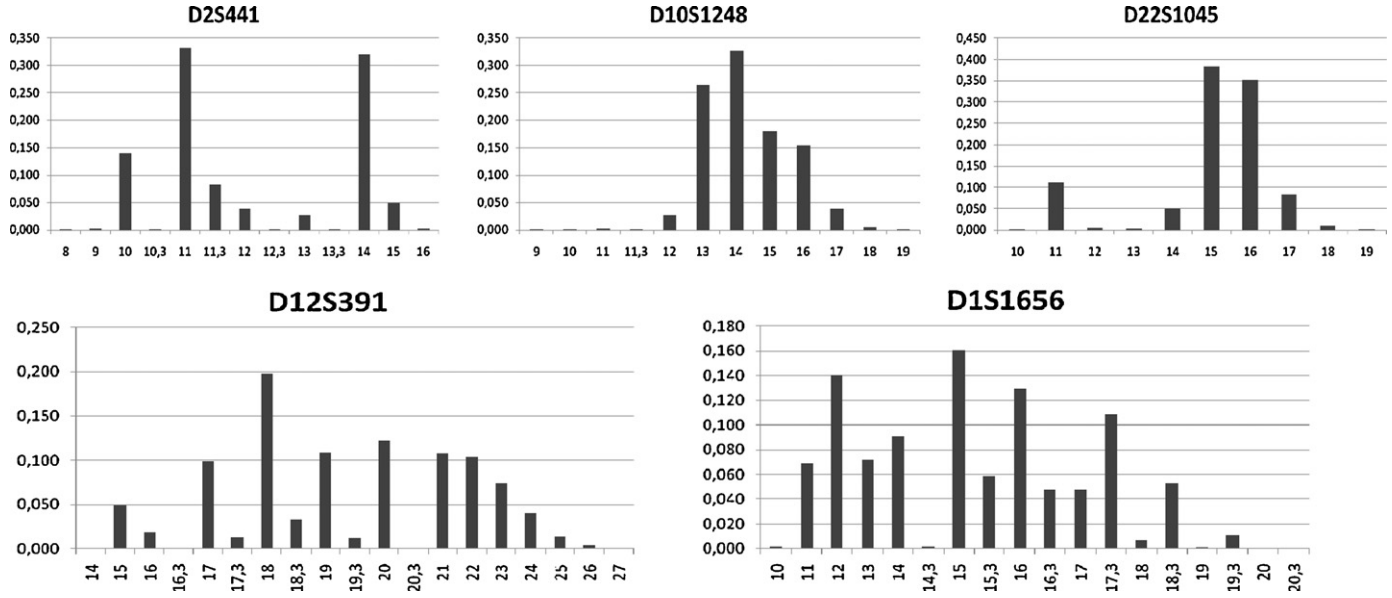

Fig. 1. Allele frequencies for the new ENFSI/EDNAP miniSTR loci in the Italian population sample. *X*-axis: allele repeats; *Y*-axis: allele frequencies.

Biosystems). The analytical data produced by the labs were collected by the Organizing Committee to check the correctness of the genetic typing.

2.6. Analysis of data

Statistical analyses were performed using Arlequin 3.0 [1].

2.7. Proficiency test

Two blind blood stains were provided by the Organizing Committee as a proficiency test.

3. Results

Fig. 1 shows the distribution of allele frequencies for the five ENFSI/EDNAP loci D1S1656, D2S441, D10S1248, D12S391 and D22S1045.

Hardy–Weinberg equilibrium was tested separately by sample ($N = 19$ samples) and by locus ($n = 5$ loci). Among the 95 independent tests performed, only three showed a $P < 0.05$, perfectly in line with the expected number of 4.75. Therefore, no evidence of deviation from HWE was observed in the data.

Heterogeneity of allele frequencies among samples was tested by computing pairwise $F(ST)$ values and by partitioning Wright's F statistics, locus by locus. None of the 171 pairwise $F(ST)$ was >0.01, testimony to a very low level of differentiation among samples. The locus by locus sample diversity measured by the F statistics was also negligible. Thus, allele frequencies were computed for all 1920 typed subjects, and these represented the Italian frequencies at these loci.

The concordance study carried out for this population study did not reveal discordant alleles for the five ENFSI/EDNAP miniSTR markers. Nevertheless, discordant typing results were seen for the CODIS loci D21S11, TH01 and for the SE33 locus. These results will be discussed elsewhere. All the groups participating in the collaborative exercise correctly typed the two blind blood stains.

Conflict of interest

None.Acknowledgements

On behalf of the Ge.F.I. Group laboratories participating in the collaborative exercise: A. Alessandrini (Università Politecnica delle Marche, Ancona), M. Alù (Università di Modena e Reggio Emilia, Modena), R. Biondo (Direzione Centrale Anticrimine della Polizia di Stato, Servizio Polizia Scientifica, Roma), I. Boschi (Università Cattolica, Roma), L. Caenazzo (Università di Padova), I. Carboni (SOD Diagnostica Genetica, Azienda Ospedaliero-Universitaria Careggi, Firenze), E. Carnevali (Università degli Studi di Perugia), F. De Stefano (Università di Genova), R. Domenici (Università di Pisa), M. Fabbri (Università di Ferrara), E. Giardina (Università di Tor Vergata, Roma), S. Inturri (Università di Torino), S. Pelotti (Università di Bologna), A. Piccinini (Università di Milano), M. Piglionica (Università di Bari), N. Resta (Università di Bari), S. Turrina (Università di Verona), A. Verzeletti (Università di Brescia).

Reference

[1] L. Excoffier, G. Laval, S. Schneider, Arlequin ver. 3.0: an integrated software package for population genetics data analysis, Evolutionary Bioinformatics Online 1 (2005) 47–50.