Forensic Science International: Genetics 7 (2013) e3–e4

Contents lists available at SciVerse ScienceDirect

Forensic Science International: Genetics

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

Letter to the Editor

Population data for 17 Y-chromosome STRs in a sample from Apulia (Southern Italy)

Dear Editor,

We evaluated allele and haplotype frequencies as well as statistical forensic parameters in a sample of 98 unrelated healthy males from Apulia region (Southern Italy) for 17 Y-STRs, using AmpFLSTR Yfiler PCR Amplification Kit (Applied Biosystems). This multiplex PCR reaction permits to amplify the loci of the European minimal haplotype (DYS19, DYS385 a/b, DYS389 I/II, DYS390, DYS391, DYS392, DYS393), the loci recommended by SWGDAM (including European minimal haplotype plus DYS438 and DYS439) and additional highly polymorphic loci (DYS437, DYS448, DYS456, DYS458, DYS635 and Y GATA H4).

Blood samples and oral swab were collected after informed consent from all individuals. Genomic DNA was extracted from peripheral blood and buccal cells transferred on FTA Cards (Whatman) by Whatman FTA purification reagents. After purification, a 1.2-mm bloodstained and buccal-swab-stained punch, containing approximately 5–20 ng DNA, was directly added to the PCR mix and amplified. The PCR amplification was carried out in a GeneAmp PCR 9700 Thermal Cycler (Applied Biosystems) according to the manufacturer's instructions for the AmpFLSTR Yfiler PCR Amplification Kit. Separation and detection of PCR products were performed on the ABI Prism 310 Genetic Analyzer (Applied Biosystems). Genotyping of each sample was carried out automatically using the allelic ladder provided with the AmpFLSTR Yfiler PCR Amplification Kit and using GeneMapper® ID software v3.2 (Applied Biosystems). After the initial genotyping, DNA samples with microvariant alleles were sequenced using Big Dye Terminator v3.1 Cycle Sequencing Kit (Applied Biosystems). The DNA control 007 included in the AmpFLSTR Yfiler PCR Amplification Kit was used as quality control template. The list of primer sequences employed for the sequencing reactions [1,2] is shown in Table 1, available as e-component.

Allele and haplotype frequencies were computed using the gene counting method. Gene diversity (GD) and haplotype diversity (HD) were calculated according to Nei [3]. Haplotype match probability (HMP), that is the probability of finding an identical haplotype in a pair of randomly unrelated males, was calculated as HMP = 1 − HD. The discriminatory capacity (DC) was determined dividing the total number of different haplotypes by the total number of individuals in the population analyzed. Pair-wise genetic distances between populations were carried out based on Φ_{st} and the significance was tested with 10,000 permutations using AMOVA tool provided by YHRD website (www.yhrd.org) [4]. A quality control was performed submitting the data to YHRD (accession number YA003730).

Several papers have been published on Y-chromosomal STR data from worldwide [5–7], including Italian [8–12] and Southern Italy [13,14] population samples. However, previously published studies regarding Apulia population were only based on 8 Y-STRs, while the present study investigated on a panel of 17 Y-STRs. We identified a total of 97 different haplotypes in a sample set of 98 unrelated individuals, with 96 haplotypes being unique and 1 occurring twice (see Supplementary Table 2). Allele frequencies and estimated values of gene diversity (GD) for each Y-STR locus are presented in Table 3, available as e-component. The lowest value of GD was observed for DYS392 (0.126), while the highest one (0.936) is presented by DYS385. The HD for the studied Y-STR set, corresponding to the chance of exclusion for unrelated males, showed a value of 0.9994, with an HMP value of 0.0006, while the overall DC was 98.98%.

Furthermore, microvariant alleles were found for the DYS458 marker, such as 17.2 (1 individual) and 18.2 (3 individuals) alleles, and in the DYS385 marker, namely the 16.3 allele (1 individual). Sequencing of these samples confirmed the insertion of two and three nucleotide pairs in these alleles, respectively. Nomenclature for these alleles was according to the International Society of Forensic Genetics (ISFG) guidelines [15,16]. In our population we also observed the 19 allele of 242 bp at DYS635 locus in one sample. This allele is not included in the allelic ladder provided with the AmpFLSTR Yfiler PCR Amplification Kit, but it has been found in other population studies [1,6,7].

When performing comparison between our population and the only Apulia sample previously submitted in YHRD (YA002985) [13,14], the AMOVA analysis detected no significant differences (p value = 0.9213). Φ_{st}-Based genetic distance was also assessed among our Apulia population and populations from South and North-West Apulia previously described elsewhere [17]. Also in this case, no evidence for significant differentiation has been detected ($\Phi_{st} < 0.08$).

Moreover, we compared Apulia Y-STR data with all populations and metapopulations belonging to the whole Mediterranean area, both Italian and non-Italian, thus-far submitted to the YHRD database. As far as concerns the comparisons with the Southern Italian populations, our analysis showed no genetic deviation ($p = 0.0664$ with San Giorgio La Molara, $p = 0.6546$ with Belvedere, $p = 0.0768$ with Trapani, $p = 0.7383$ with Catania) [18,19]. By contrast, AMOVA analysis indicated significantly differences ($p < 0.05$) with respect to all Northern Italian populations (Udine, Biella, La Spezia, Modena, Ravenna, Marche and North Sardinia) [18,20–23]. Regarding the non-Italian populations belonging to the Mediterranean basin, AMOVA analysis showed that our Apulia sample had significant differences with all the populations included in YHRD (Croatia, Macedonia, Albania, Greece, Turkey, Israel, Libya, Tunisia, Algeria, Morocco and Spain) [6,7,24–28]. The geographical location of each population included in the comparison is shown in Supplementary Figure 1.

This paper follows the guidelines suggested for publication of population data in Forensic Science International [29].

1872-4973/$ – see front matter
http://dx.doi.org/10.1016/j.fsigen.2012.08.003

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at http://dx.doi.org/10.1016/j.fsigen.2012.08.003.

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20 January 2012