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Forensic Population Genetics – Letter to the Editor

Genetic population studies on 15 NGM™ STR loci in central Poland population

Dear Editor,

We investigated genetic polymorphism of 15 STR loci (D10S1248, D22S1045, D2S441, D1S1656, D12S391, D2S1338, D3S1358, D8S1179, D16S539, D18S51, D19S433, D21S11, FGA, TH01, vWA) included in the NGM™ amplification kit in a sample of 400 unrelated, adult individuals (189 females and 211 males) born in the Lodz region of central Poland.

Genomic DNA was extracted from buccal swab samples with the AX Sherlock Kit (A&A Biotechnology, Poland) according to manufacturer's protocol. The concentration of extracted DNA was determined with the Qubit™ Quantitation Platform (Invitrogen, USA) and 7500 Real-Time PCR System with HID Real-Time PCR Analysis Software v.1.0 (Applied Biosystems, USA).

The amount of 0.5–1 ng target DNA template was amplified in a multiplex-PCR reaction using AmpF*l*STR® NGM™ PCR kit and GeneAmp 9700 PCR System (Applied Biosystems, USA).

PCR products were detected in a five-dye detection system by capillary electrophoresis in a 3500 Genetic Analyzer (Applied Biosystems, USA). The results were analyzed with GeneMapper® ID-X Software v.1.2 (Applied Biosystems, USA) using LIZ 600 v.2 size standard with reference allelic ladder and male 007 DNA control template provided with NGM™ kit according to the recommendations of the DNA ISFG Commission [1].

Estimations of allele frequency for each autosomal locus of NGM™ kit [2] were determined with direct counting method. Obtained allele frequencies results are presented in Supplementary Table 1. Population genetics statistical parameters such as Polymorphism Information Content (PIC), Power of Discrimination (PD), Power of Exclusion (PE), Matching Probability (MP) and Typical Paternity Index (TPI) were calculated using PowerStats spreadsheet v. 1.2 [3]. Statistical analysis performed with GDA software included: expected Heterozygosity (H_{exp}), observed Heterozygosity (H_{obs}) and Hardy–Weinberg equilibrium deviation for each analyzed locus [4]. Obtained results are summarized in Supplementary Table 2. No deviations from HWE were observed for 14 STR of 15 NGM loci, except D3S1358 locus. No excess of homozygotes was observed at that locus. After applying Bonferroni's correction for multiple testing (significance value $p = 0.0033$) to investigated loci, D3S1358 locus deviation appeared statistically significant (p-value = 0.002), what may be explained by limited population size.

The set of 15 STRs characterizes a very high combined power of discrimination and combined power of exclusion equaled 0.9999999999999999998 and 0.9999997 respectively. Among five new loci included in NGM™ kit (D10S1248, D22S1045, D2S441, D1S1656, D12S391) for two of them were found very high values of power of discrimination (D1S1656: 0,979 and D12S391: 0,973) and observed heterozygosity (0,888 and 0,868, respectively), what may be crucial in solving some forensic cases and chimerism analysis. It has been confirmed by our chimerism studies carried out by the samples obtained from patients after hematopoietic stem cell transplantation (data not published yet). The probability that two randomly selected individuals from the region of central Poland population have exactly the same 15-loci genetic profile is 2.1×10^{-19}. Obtained results for analyzed loci set stay in agreement with other researchers' results [5,6].

Observed allele frequencies in analyzed population from region of Lodz ($n = 400$) were compared to other Polish population data – from Cracow in southern Poland ($n = 154$) [5]. Pairwise interpopulation comparisons were performed by the RxC software (G. Carmody, Canada) using Monte Carlo simulations to calculate the statistical significance of the X^2 two-way contingency tables. The correction for the number of interpopulation comparisons at an adjusted significance level was made. Interpopulation comparison revealed statistically significant differences between Lodz and Cracow populations in D3S1358, D21S11 and D1S1656 loci ($p < 0.001$) even after applying Bonferonni's correction for the number of comparisons.

The performed evaluations of the usefulness of the human autosomal STR markers by analysis of statistical parameters (PD, PE, PIC, MP, TPI) have confirmed their significant informativeness and thus their usefulness in forensic genetics studies. Extremely high combined power of discrimination value and sensitivity of NGM™ loci set seems to be very important for data sharing and analyzing challenging samples. Investigated markers can be also applied for detection of chimerism within patients after hematopoietic stem cell transplantation, what will be the aim of our further report.

Our laboratory participates in GEDNAP and Polish Society for Forensic Genetics & Criminology (www.ptmsik.pl) proficiency tests for forensic DNA typing certificates.

This paper follows the guidelines for publication of population data requested by the journal [7].

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.fsigen.2011.10.004.

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