Forensic Science International: Genetics 9 (2014) e20–e21

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Letter to the Editor

Y-STR haplotype diversity in Punjabi population of Pakistan

Dear Editor,

We studied the genetic diversity of 17 Y-chromosomal STR loci (DYS19, DYS385a/b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 and Y-GATA H4) in a sample of 300 genetically unrelated males from Punjabi population of Pakistan.

DNA was extracted from blood stains collected on blotting paper (Whatman, Kent, UK) using Phenol chloroform extraction method [1] and quantified using Quantifiler® Human DNA Quantification Kit using ABI 7500 Real-time PCR System. Y-chromosomal STR loci were amplified using AmpFLSTR® Yfiler™ PCR kit with GeneAmp® 9700 PCR System (Life Technologies Inc.) according to the manufacturer's instructions. Amplified products were analyzed by using the ABI PRISM® 3100 Genetic Analyzer according to the manufacturer's recommended protocol. ABI GeneScan® 500-LIZ™ was used as internal size standard along with ABI Hi-Di™ Formamide. The data were analyzed using ABI GeneMapper ID v3.2. Alleles were designated according to the latest recommendations of the International Society for Forensic Genetics [2].

Haplotype and Allele Frequencies were estimated by simple counting using a spreadsheet program. Discrimination Capacity (DC) of the haplotypes was calculated as $DC = H/n$, where H is the number of different haplotypes and n is the number of total samples. Estimation of haplotype and gene diversities together with intra- and inter-population indices was computed using ARLEQUIN v3.5 [3]. Analysis of molecular variance (AMOVA) was calculated using the AMOVA tool available at YHRD website (www.yhrd.org/Analyse).

Total 274 different haplotypes were observed, in which 254 were unique while 20 were observed more than once (Supplementary Table S1). The discrimination capacity was 91.33% while unique haplotypes accounted for 92.7%, showing that these loci are useful for paternity testing and personal identification in the Punjabi population. Locus DYS385 showed the highest diversity (0.9171) while locus DYS391 showed the lowest diversity (0.4137).

Supplementary material related to this article can be found, in the online version, at doi:10.1016/j.fsigen.2013.12.004.

Punjab (*Persian*; five waters), is the largest province of Pakistan by population accounting for 56.2% of the country's population of 180 million (www.census.gov.pk). It is believed that urban civilization existed in the region before the Aryans invasion in around 2500 BC. Occupying a strategically key location, the region has remained part of many empires and civilizations during the course of history. It remained an important hub of Mughal Empire followed by a brief rule by the Sikhs before it fully came under the British Rule and later became part of Pakistan [4]. We compared our data to that of 10 other populations: Afghanistan [Pathan] [5], Ahvez, Iran [Arab] [6], Dhaka, Bangladesh [Bangladeshi] [7], Punjab, India [Balmiki] [8], Kashmir, India [Saraswat Brahmin] [9], Punjab, India [Saraswat Brahmin] [9], Beijing, China [Han] [10], Xi'an, China [Han] (YHRD Accession Nr. YA003671), Eastern Mongolia [Khalkh, Buryat, Dariganga] (YHRD Accession Nr. YA003736), Khyber Pakhtunkhwa, Pakistan [Yousafzai Pathan] (YHRD Accession Nr. YA003748).

The Rst values for measuring the genetic distances between 11 populations are presented in Supplementary Table S2. The results demonstrate that Punjabi population of Pakistan formed a close cluster with the Sarswat Brahmin population of Indian Punjab ($p = 0.2266$ and $Rst = 0.003$). Apart from this, all the other populations, including the Balmiki population which was reported from the Indian Punjab as well as Sarswat Brahmin population of Kashmir, showed significant difference from the Punjabi population of this study.

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As shown in the Multidimensional Scaling plot (MDS) in Supplementary Figure S1, the Punjabi population of Pakistan tends to be closer to Bangladeshi population, Indian (Punjab), Kashmiri and then Iranians. Punjabi population is distant from the Pathan populations of Afghanistan as well as Yousafzai. Moreover, populations of Mongolia, Balmiki and Chinese were at a great distance from Punjabi population of Pakistan.

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We compared our data with 12 other Pakistani populations for which Y STR data is available for 10 loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438 and DYS439) [11]. As shown in Supplementary Figure S2, number of alleles reported for these 12 populations is only slightly different from that of the Punjab.

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The data was submitted to YHRD (www.yhrd.org) and received the accession number: Punjab, Pakistan [Punjabi], $n = 300$, YA003781. The recommendations of the ISFG were strictly adhered to in this study [12]. We have followed the guideliness of this journal for publication of population data [13].

Acknowledgements

We are thankful to all the people who participated in this study by donating their DNA samples. We would also like to appreciate the co-operation of genotyping laboratory as well as all the support staff of CEMB and CAMB.

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22 October 2013