Forensic Science International: Genetics 7 (2013) 214–216

Contents lists available at SciVerse ScienceDirect

Forensic Science International: Genetics

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

Short communication

Haplotype diversity of 17 Y-STR loci in a Chinese Han population sample from Shanxi Province, Northern China

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ARTICLE INFO

Article history:
Received 3 April 2012
Received in revised form 13 June 2012
Accepted 6 October 2012

Keywords:
Y-chromosomal STR
Shanxi Province
Haplotype
Multidimensional scaling analysis

ABSTRACT

The distribution of 17 Y-chromosome STR loci DYS456, DYS389I, DYS390, DYS389II, DYS458, DYS19, DYS385a/b, DYS393, DYS391, DYS439, DYS635, DYS392, Y-GATA-H4, DYS437, DYS438, and DYS448 haplotypes was determined in a population sample of 222 unrelated Chinese Han from Shanxi Province, Northern China. A total of 219 haplotypes were observed, and of these, 216 were unique, while 3 were found two times. The overall haplotype diversity was 0.9999 and the discrimination capacity was 0.9865, indicating a high potential for differentiating between male individuals in this population. Comparison analysis via Analysis of Molecular Variance (AMOVA) and construction of MDS plot revealed that Shanxi Han sample clusters with Chinese origin populations and stands far apart of the non-Chinese populations, justifying the establishment of local databases in Shanxi Han population for any future forensic and genetic epidemiology efforts in this region.

1. Population

Blood samples were randomly collected from 222 unrelated healthy male individuals of Chinese Han population living in Shanxi Province, Northern China. All participants signed the informed consent and provided the information about birthplace, parents and grandparents at the same time. Their ancestors had lived in the region for at least three generations. Shanxi Province is one of the oldest territories in the current country of China with a history leading back to before the Spring and Autumn Period (722–403 BC) when it was the location of the military powerful state of Jin. Under the Han and Tang Dynasties already, Shanxi was a territory or Province of China. As a result of its mountainous nature, the Province is rich in mineral resources, and since the 1930s has been developed as the Coal (and Iron) producing heart of China. Shanxi has a population of over 35 million (year of 2010), including its minority ethnic population, and the Han ethnicity makes up almost the entire population with 99.75%.

2. DNA extraction

Genomic DNA was extracted using the Chelex-100 method as described by Walsh et al. [1].

3. Amplification

17 Y-STR marker (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS438, DYS439, DYS437, DYS448, DYS458, DYS456, DYS635, and Y-GATA-H4) were co-amplified using an AmpFLSTR Y-filer kit (Applied Biosystems, USA) [2]. PCR amplification reactions were carried out using a GeneAmp PCR system 9700 (Applied Biosystems, USA) following the protocol provided by the manufacturer.

4. Electrophoresis and typing

The amplified products were separated by capillary electrophoresis on ABI Prism1 3130 Genetic Analyzer (Applied Biosystems, USA) using GeneScanTM-500 LIZTM internal size standard. The sample run data were analyzed together with an allelic ladder and positive and negative controls using GeneMapper ID Software Version 3.2 (Applied Biosystems, USA). The updated recommendations of the DNA Commission of the International Society of Forensic Genetics for analysis of Y-STR systems were followed [3].

5. Analysis of data

Allelic frequencies were estimated by direct gene-counting. Gene and haplotype diversities were calculated according to the formula by Nei [4]. The discrimination capacity was calculated as the proportion of different haplotypes in the sample. Pairwise values of Rst were calculated to measure the genetic distance

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http://dx.doi.org/10.1016/j.fsigen.2012.10.004

corresponding to complete 17-marker haplotypes (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a, DYS385b, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, Y-GATA-H4) of our population and compared with 11 other published data or data from neighbouring countries submitted to Y-STR haplotype database (YHRD), using ARLEQUIN software Version 3.1 [5]. To illustrate the relationship between populations based on pairwise Rst, a multidimensional scaling (MDS) plot was created by using SPSS 15.

6. Quality control

Our laboratory has participated in the Y-STR haplotype reference database (YHRD) quality assurance exercise in 2009 typing the YHRD core loci as well as additional loci DYS437, DYS448, DYS456, DYS458, DYS635 and Y-GATA-H4. The Y-STR haplotype data were contributed to the Y chromosome STR haplotype reference database (http://www.yhrd.org), with the accession number YA003589 and population ID YP000622.

7. Results

Supplementary Table S1 summarizes the allele frequencies of 17 Y-STR loci, while Supplementary Table S2 lists the haplotypes of 222 unrelated Chinese Han samples from Shanxi Province. The Rst values calculated to measure genetic distances between 17 Y-STR haplotypes of 11 neighbour populations ($n = 5269$) with the statistical significance were calculated in supplementary Table S3.

8. Other remarks

Among the 17 markers analyzed, DYS385 and DYS391 were calculated to be the highest (0.9755) and lowest (0.3894) values for gene diversity, respectively (Table S1). A total of 219 different haplotypes were identified from 222 unrelated male individuals, of which 216 were unique and 3 were found in 2 individuals (Table S2). The overall haplotype diversity was calculated as 0.9999 with a discrimination capacity of 0.9865. The results indicate that these 17 Y-STR loci are useful genetic markers for forensic personal identification and paternity testing in the Chinese Han population.

We also compared our data of extended minimal haplotypes (minimal haplotype + DYS438 and DYS439) with YHRD database, Release 39, which currently includes 72,171 extended minimal haplotypes over 519 populations. One hundred and seventy-four (79.45%) haplotypes detected in Shanxi population are in zero matches in YHRD with extended minimal haplotypes (ExHt) database. Ht15 is found to match most frequently in YHRD ExHt database with a hit of 44 of which 35 with East Asian Metapopulation. Ht15 was followed by Ht176 and Ht93. For Ht176, out of 20 hits, 19 hits were observed with East Asian Metapopulation. Whereas for Ht93, 18 hits were observed with East Asian out of 19 hits.

For having extensive illustration of the genetic relation, the studied data were compared via AMOVA on the same 17 Y-STR loci set with data from 11 reference populations (published and referenced in the YHRD). Namely 119 Han Chinese individuals residing in South China [6], 207 Han Chinese individuals from Beijing (YHRD, Accession # YA003470), 203 Han Chinese individuals from Zhejiang [7], 200 Han Chinese individuals from Taiwan [8], 1079 Japanese individuals [9], 331 Chinese individual from Malaysia [10], 334 Malay individuals from Malaysia [10], 315 Indian individuals from Malaysia [10], 1021 Korean individuals from South Korea [11], 481 Brazilian individuals [12], and 216 Bangladeshi individuals [13] with the statistical significance determined by a permutation test

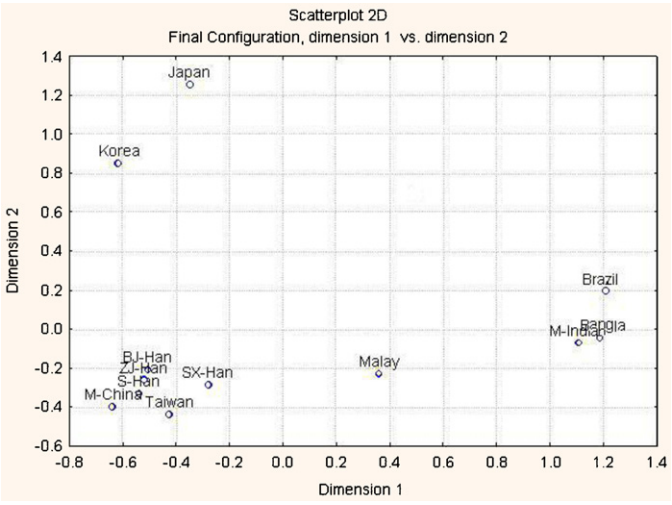

Fig. 1. Multi-dimensional scaling (MDS) plot of the Chinese Han population from Shanxi and 11 reference populations, from pairwise Rst values. Stress value = 0.002. Acronyms are as follows: SX-Han, Han Chinese population from Shanxi; BJ-Han, Han Chinese population from Beijing; ZJ-Han, Han Chinese population from Zhejiang; S-Han, Han Chinese population from South China; Taiwan, Han Chinese population from Taiwan; M-China, Malaysia Chinese; Malay, Malaysian; M-Indian, Malaysia Indians; Bangla, Bangladeshis; Brazil, Brazilians; Japan, Japanese; Korea, South Koreans.

(10,000 replicates, Table S3). AMOVA analysis showed that 88.56% of the variation was found within populations, whereas 11.44% was among populations (fixation index $FST = 0.1144$, $P = 0.00000$). Pairwise analysis showed no significant differences ($P > 0.05$) in the comparison of Shanxi Han and Beijing Han (Rst = 0.00374). With other Chinese origin samples from South China, Zhejiang, Malaysia, and Taiwan, although significant, low Rst values were obtained (0.01377, 0.01521, 0.02091 and 0.03059, respectively). In the comparison with the remaining populations, highly significant distances were observed ($P = 0.00000$).

The MDS plot (Fig. 1) structured from Rst distance matrix shows that Shanxi population along with Beijing Han, Zhejiang Han, South Han Chinese, Malaysian Chinese and Taiwan Chinese populations form a conspicuous cluster standing far apart from other Asian populations. As demonstrated in the MDS plot, Bangladeshi and Indian individuals from population had a close genetic relationship (Rst = 0.02736). This reflects that culture trait may be related to migration and marriage. Gene flow is influenced by culture background in Chinese populations. Similarity of culture is easier to result in genetic compatibility.

Haplotypes of 17 Y-STR loci in our population were contributed to the Y-STR haplotype reference database (http://www.ystr.org). This study was followed the guidelines for publication of population data requested by the journal and the DNA Commission of the International Society of Forensic Genetics [3,14].

Acknowledgments

We thank all sample donors for their contributions to this work and all those who helped with sample collection. This study was supported by The National Natural Science Foundation of China (NSFC) (No. 81172902), and The Program for New Century Excellent Talents in University (No. NCET-10-0773) and the New-Star Program of Science and Technology of Beijing Metropolis (2010B073), and the Program for Young Innovative Research Team in China University of Political Science and Law.

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.2012.10.004.

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