

Y-Str Haplotyping For The North Indian Population Of India.

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Abstract

In 200 males from the Northern Indian population, seventeen Y-STR loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 and Y-GATA-H4). The AmpFISTR® Yfiler® Kit was used to conduct multiplexed PCR amplification of the 17 Y STR loci. Allele frequencies and population genetic diversity were determined. The AMOVA tool was used to compare the haplotype data collected in the analysis to the Y-STR haplotypes reference database (YHRD, <http://www.yhrd.org>) and to previously reported population data, and the results were visualized in two-dimensional multidimensional scaling (MDS) plots. A total of 200 different haplotypes were discovered, with 145 of them being distinct. Twenty-five haplotypes were discovered twice, one was discovered three times, and one was discovered four times. The discriminating ability and haplotype diversity were found to be 0.9980 and 0.8499, respectively. At the loci, gene diversity ranged from 0.398 to 0.785. The Genotype Diversity (GD) at the studied loci ranged from 0.399 to 0.788, with 0.871 being found at the DYS385a/b locus. When compared to populations from other parts of the Indian subcontinent and population data from other countries, the population of Northern India was found to be significantly different ($p < 0.05$).

Keywords: Northern India, 17 Y STR loci, Allele frequencies, Y-STR haplotypes, population genetic diversity

Introduction

Early Y-chromosome short tandem (STR) markers were discovered during cloning experiments or were obtained from the Genome database in silicon (GDB) [1-10]. These markers, for example, include the nine loci that comprise the "minimal haplotype" (MHT), which remains the centre stage of all Y-STR packaging in current forensic applications while also representing a rather heterogeneous and random range of markers with different population genetic properties [11-15]. At the same time, a sequence has been carried out of the whole euchromatic region of the human Y-chromosome and a systematic quest for potentially useful Y-STRs has now become practicable [15] [19]. New Y-STRs were identified and combined in a so-called fast-moving (RM) marker-group [16], 13 with the highest mutation rate, in a recent study by Ballantyne et al. At least one RM Y-STR mutation may also be resolved between 50% and 100% of related men in the same study (up to 20 meioses apart) [17-18].

These findings show that low-level haplotype sharing among patrilineal families is generally linked to RM Y-STR combinations, overcoming a forensic evidence constraint in the form of Y-

STR [20-25]. The multi-copy structure of some of the most mutable Y-STRs makes incorporating RM approaches into forensic cases challenging and often inefficient [26]. The Indian Subcontinent is rich in a diverse demographic mix, with a population that ranges [27] between a few hundred and a million people (Census of India, 2011) [28]. Many indigenous populations have a significant degree of genetic variation, according to genetic populations based on polymorphous Y STR DNA [27][29][30]. This research examined population genetic variation and allele frequencies in a North Indian population on 17 STR loci, with the aim of adding them to a forensic database.

Methods and Protocol

Ethical Clearance

The Ethics Committee approved the collection of 200 blood samples from 200 healthy unrelated males in North India using a standard protocol on Whatman FTA Cards (GE Healthcare, USA). FTA Cards were held at RT until they were processed further.

Data generation and sample processing

Automate Express extracted DNA from the above FTA Cards (using a Harris Uni-Corn disposable punch 2.00mm) (Applied Biosystems, now part of Thermo Fisher Scientific Inc., Foster City, CA) [31-40]. The Quantifiler® DNA Quantification Kit was used for quantification of the isolated DNA using the Real-Time PCR ABI 7500 HID (Applied Biosystems, now part of Thermo Fisher Scientific Inc., Foster City, CA) [41] as per the manufacturer's suggested protocol.

The PCR program was carried out on an Applied Biosystems Veriti Thermal Cycler with the AmpFlSTR® Yfiler® Kit, as recommended by the manufacturer. The ABI 3500 Genetic Analyzer (Applied Biosystems) was used to separate and detect DNA fragments on a G5 matrix set using matrix standards DS-33 [42]. As an internal size standard, the Gene Scan™ 500 LIZ size standard (Applied Biosystems) was used. GeneMapper Analysis Software v3.5 was used to compare the amplified DNA fragments to those of an allelic ladder provided with the package to determine allele designations (Applied Biosystems) [43]. For allele designation, a peak detection threshold of 50 RFUs was used [44]. Both procedures were carried out in accordance with the laboratory's internal controls and kit controls [45]. Quality Assurance The YHRD's quality management exercise was successful (www.yhrd.org). Internal laboratory control standards and kit controls were also used [46-50].

Management of data and statistical analysis

DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS 385a/b, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 and Y-GATA-H4 are among the 17 Y-STR loci studied. GenAlEx 6 was used to quantify allele frequencies [51]. Haplotype was calculated using Haplotype Analysis v1.04 [52]. The formula for calculating gene diversity (GD) was:

$$GD = \frac{n}{n-1} \left[1 - \sum_{i=1}^n P_i^2 \right]$$

where P_i represents the frequency of the i th allele and n represents the number of samples examined [24]. The haplotype diversity (HD) was estimated using the following formula:

$$HD = \frac{n}{n-1} \left[1 - \sum_{i=1}^n X_i^2 \right]$$

where X_i represents haplotype frequency [53-55]. To find out the genetic relationship, the observed haplotypes in the study were compared with other countries population database based on F_{st} values using the AMOVA online tool Y-STR haplotypes reference database (YHRD, <http://www.yhrd.org>) and visualized in two-dimensional multidimensional scaling (MDS) plots.

Count of DYS390

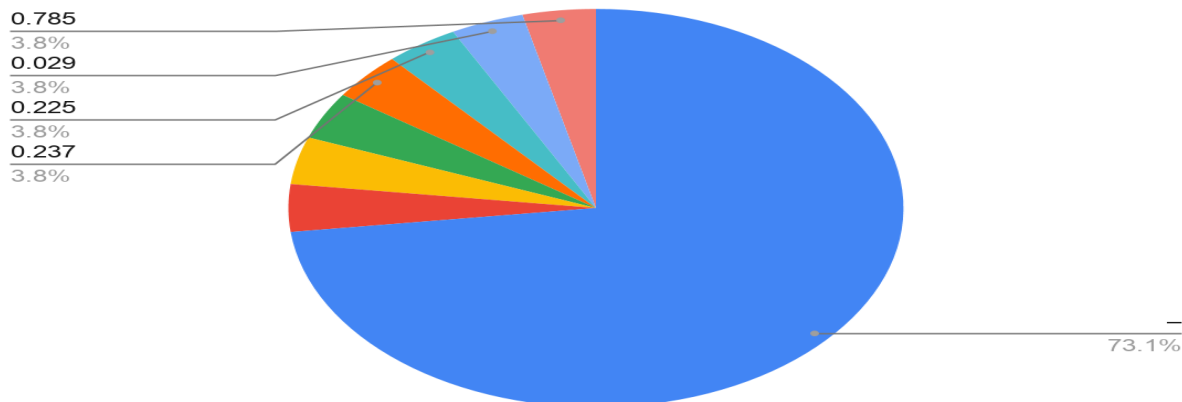


Figure 1. Allele frequencies and gene diversity values of 17 Y- STR loci in North Indian Population. The table shows Allele Frequencies for Each Y STR except DYS390a/b for which Genotype frequencies are reported.

*GD, Gene Diversity.

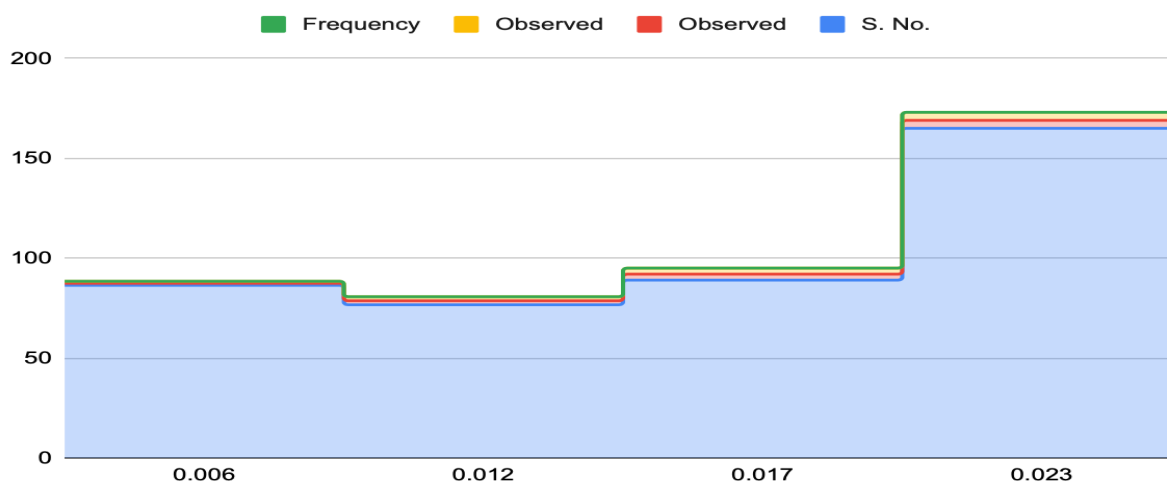


Figure 2. Shows Allele Frequencies for each Y STR locus Except DYS385a/b, for which Genotype Frequencies are Reported. X-axis is the frequency and the y-axis is the haplotype of Y-STR.

Results

A total of 200 unrelated individuals were typed for 17 Y- STR Loci. The allele frequencies and gene diversity (GD) are shown in Figure 1. The GD at the studied loci ranged from 0.399–0.788 and the Genotype Diversity at the locus DYS385a/b was found to be 0.871 (figure 1). A total of 200 different haplotypes were observed, out of which 145 were unique, 25 haplotypes were observed twice, one was observed three times and one haplotype was observed four times (figure 2). Haplotype diversity and discriminating capacity were found to be 0.9980 and 0.8499, respectively (figure 2). The studied Northern Indian Population showed a significant difference MDS Plot (Figure 3) ($p < 0.05$) with all the compared populations reported from [other countries](https://yhrd.org) [Denmark, Estonia, Finland, Iceland, Ireland, Latvia, Lithuania, Sweden and the United Kingdom (<https://yhrd.org>)].

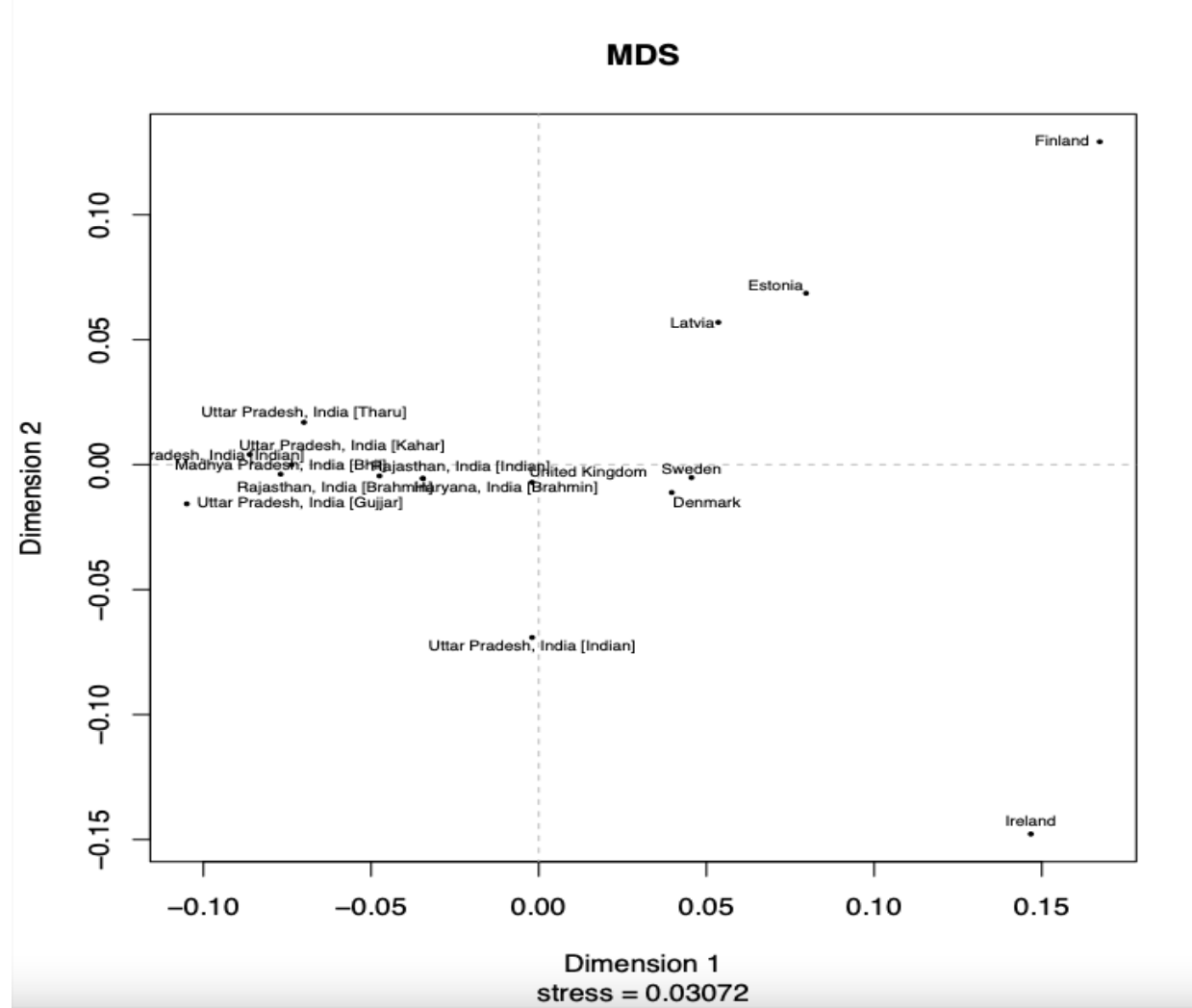


Figure 3. MDS plot showing the relationship of the Studied North Indian Population with Population Data from Denmark, Estonia, Finland, Iceland, Ireland, Latvia, Lithuania, Sweden and the United Kingdom (<https://yhrd.org>).

Conclusion

Although when compared to populations from a sample size of 200 (40 from each state such as Rajasthan/U.P./M.P./Haryana/Punjab), The studied Northern Indian population showed a significant difference (Figure 3) ($p < 0.05$) with all the compared populations reported from other countries. This study's population data can be used in forensic, anthropological, and demographic studies.

Declaration of interest

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

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