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POPULATION DATA

Population genetic data for ten miniSTR loci in the Sri Lankan population

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Abstract Allele frequencies and forensically important parameters of ten autosomal miniSTR loci, D1S1677, D2S1776, D10S1248, D11S4463, D12SATA, D14S1434, D17S974, D18S853, D20S482, and D22S1045, were obtained for 278 unrelated adults from the Sri Lankan population. The combined power of discrimination and probability of exclusion was found to be 0.999999999621539 and 0.9979620, respectively. No significant deviations from Hardy–Weinberg equilibrium were observed except for D20S482 which conformed to HW expectations only after the application of a Bonferroni correction. The study suggests the potential use of these miniSTRs as a supplement or as a stand-alone STR marker system for the analysis of highly degraded evidence in Sri Lanka.

Keywords Degraded DNA · Miniaturized short tandem repeats (miniSTR) · Sinhalese · Sri Lankan Tamils · Sri Lankan Moors

The use of STR markers in the identification of individuals plays a significant role in forensic investigations. However, degraded DNA hinders successful retrieval of information leading to the loss of typing data. The DNA degradation is further influenced by tropical climatic conditions as prevail in Sri Lanka. Thus, establishing miniSTR typing capabilities for Sri Lankan forensic casework is of great importance.

We investigated ten miniaturized Short Tandem Repeat (miniSTR) loci out of 26 non-CODIS miniSTRs published by the National Institute of Standards, USA (NIST). Samples in this study were collected from 278 healthy, unrelated volunteers belonging to the three major ethnic groups (Sinhalese, Sri Lankan Tamils, and Sri Lankan Moors) which comprise over 95% [1] of the Sri Lankan population.

Blood samples were collected from a total of 278 unrelated healthy individuals upon obtaining informed written consent. Genomic DNA was extracted from whole blood by Chelex-100 DNA extraction protocol [2]. Singleplex PCR reactions were performed for genotyping each of the ten miniSTR loci using the primers originally described by Hill et al. [3]. Each singleplex PCR reaction was performed in a total volume of 25 μl containing 10 ng of genomic DNA, in a GeneAmp® PCR System 2700 (Applied BioSystems, Foster City, CA, USA) thermal cycler following reaction conditions described in Hill et al. [3]. Each PCR reaction was carried out with K562 reference DNA (Promega, USA) as a positive control and ddH_2O as a negative control. Amplified PCR products were separated on a Gibco BRL S2001 electrophoresis apparatus, followed by silver staining with the GenePrint® STR Systems Silver Stain Detection kit [4] (Promega, USA) as per manufacturer's recommendations.

The allelic ladders were generated by combining DNA from up to ten individuals and characterizing their PCR products by DNA sequencing and by co-migration with K562 DNA reference alleles [5]. The nomenclature of the alleles

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was done as per the recommendations of the DNA Commission of the ISFG [6].

Statistical evaluation of allelic data was performed using PowerStats V1.2 software [7]. Allele frequency distribution and forensically important parameters such as power of discrimination (PD), power of exclusion (PE), and polymorphism information content (PIC) were obtained for the ten candidate miniSTR loci and a listed in Supplementary Table 1. Hardy–Weinberg and linkage equilibrium for each miniSTR locus was verified with Arlequin 3.5.2.2 software [8]. Nine out of ten miniSTR loci did not show significant deviations from the Hardy–Weinberg equilibrium (HWE) for the tested population, and D20S482 conformed to HWE expectations only after the application of Bonferroni's correction. Allele ranges observed for the ten miniSTR marker panel for the Sri Lankan population ranged from 58 base pairs to 161 base pairs. MiniSTR loci D1S1677, D10S1248, D11S4463, D12SATA, D14S1434, D17S974, D18S853, D20S482, and D22S1045 generated PCR fragments less than 120 base pairs, while D2S1776 extended to 161 base pairs. MiniSTR markers D1S1677, D2S1776, D10S1248, D11S4463, D12SATA, and D14S1434 showed a relatively high degree of polymorphism with observed heterozygosities H(ob) > 0.7, D17S974, D18S853, and D22S1045 showed a moderate degree of polymorphism with the observed heterozygosities H(ob) > 0.65, and D 20S482 showed the lowest observed heterozygosity of 0.59. The combined power of discrimination (CPD) and the combined power of exclusion (CPE) for the ten studied loci were 0.999999999621539 and 0.9979620, respectively. Significant linkage disequilibrium was observed between miniSTR loci D2S1776-D11S4463 (P = 0.02475) and D20S482-D22S1045 (P = 0.00851) prior to the Bonferroni's correction and is summarized in Supplementary Table 2. A population comparison was done based on pairwise Fst values calculated with Arlequin 3.5.2.2 software from the allele frequencies of the ten miniSTR loci for the three Sri Lankan ethnic groups and for the four populations in the USA for which data was available [3]. A multidimensional scaling plot of pairwise Fst values depicted in Fig.1 was derived using XLSTAT 2016.1 software [9]. As expected, the three Sri Lankan ethnic groups appeared to have the closest genetic affinity with the US Asian populations. US Hispanic, US Caucasian, and African American populations were placed at a greater distance on the plot.

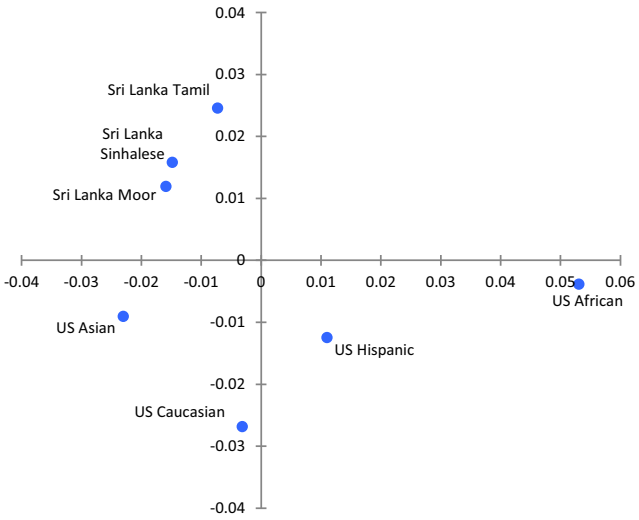

Fig. 1 Multidimensional scaling plot of population pairwise Fst for three Sri Lankan ethnic groups (Sinhalese, Sri Lankan Tamils, and Sri Lankan Moors) and for four US populations (Caucasian, Asian, Hispanic, and African American)

The ten miniSTR loci have shown relatively high power of discrimination for the Sri Lankan population which suggests its potential use in forensic and human parentage analysis in Sri Lanka.

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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