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POPULATION DATA

Genetic polymorphism analysis of 23 STR loci in the Tujia population from Chongqing, Southwest China

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Abstract

To evaluate the applicability of 23 autosomal STR loci (D10S1248, D11S4463, D12ATA63, D14S1434, D17S1301, D18S853, D1GATA113, D1S1627, D6S1017, D20S1082, D20S482, D17S974, D22S1045, D1S1677, D2S1776, D2S441, D3S4529, D4S2408, D9S1122D5S2500, D6S474, D18S51, D9S2157) included in DNA Typer™ 25 Kit for individual identification and parentage testing, allele frequencies and forensic efficiency parameters were first obtained from healthy, unrelated 506 Chongqing Tujia individuals. A total of 1012 alleles were identified in 23 STR loci, and allele frequencies ranged from 0.001 to 0.5761. The combined power of discrimination (CPD) and the combined power of exclusion (CPE) of the 23 STR loci were 0.99999999999999999999753 and 0.99999967, respectively. These results suggested that 23 autosomal STR loci could be used as an effective tool for forensic application in Chongqing Tujia population. Comprehensive comparisons were conducted based on the analysis of genetic distance, principal component analysis (PCA), multidimensional scaling plot (MDS), and phylogenetic tree to explore the interpopulation genetic relationship. Our results revealed that Chongqing Tujia keeps the more relatively genetic similarity with Hunan Han, Hubei Tujia, and Sichuan Han, which could be interpreted by that those populations were originated from the same ethnic ancestor or genetic communication were happened in adjacent areas.

Keywords STR · Chongqing Tujia · Genetic polymorphism · Evolutionary relationship

Introduction

Chongqing, situated in southwest of China, is the largest municipality. The population of Chongqing is primarily comprised of Han (94.2%), with minorities of Tujia (4.56%), Miao. According to the 6th national census, the total population of Tujia reached 8 million, which is the 8th largest Chinese ethnic minority. The Tujia people mainly reside in the Wuling Mountains, which occupy the border zone of Chongqing, Hunan, Hubei, and Guizhou. As previous literatures claimed, the history of Tujia can be traced to the ancient Ba which occupied the modern Chongqing around 2500 years ago. The Tujia own their spoken language which belongs to the Sino-Tibetan language group and has grammatical and phonological similarities with Nuosu.

Short tandem repeats (STR), a highly polymorphic DNA marker, were commonly used in forensic application and scientific research for its wide distribution, high polymorphism, easy detection and stable heritability. Since the accuracy of the personal identification and paternity test was directly affected by the mutation rate and different gene frequencies, it is significantly important to obtain the STR polymorphism of the local population, as well as explore the genetic structures among different groups. Moreover, reference data for studies of population genetic relationship and evolution process could be enriched.

The present study aimed to evaluate genetic polymorphism of Typer™ 25 Kit (Institute of Forensic Science, Ministry of Public Security, P.R. China) [1, 2] in Chongqing Tujia population, the novel system containing 23 autosomal STR loci (D10S1248, D11S4463, D12ATA63, D14S1434, D17S1301, D18S853, D1GATA113, D1S1627, D6S1017, D20S1082,

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D20S482, D17S974, D22S1045, D1S1677, D2S1776, D2S441, D3S4529, D4S2408, D9S1122, D5S2500, D6S474, D18S51, D9S2157) as well as two sex determining loci (DYS391 and amelogenin). Allele frequencies and parameters were determined to estimate the effectiveness of this new multiple PCR system in forensic application. Population comparisons and evolutionary analysis were performed to demonstrate the relationship between geographic location and genetic inheritance.

A total of 506 (312 males and 194 females) blood stains on the FTA card of Tujia individuals were collected from Chongqing Municipality with their written informed consent. The genomic DNA were amplified directly without extraction. Direct amplification was conducted with Typer™ 25 Kit on GeneAmp PCR System 9700 Thermal Cycler, according to the manufacturer's instructions. Electrophoresis data and alleles identification were analyzed by GeneMapper ID-X v.1.4. Our study was undertaken in Genetic Laboratory of Institute of Forensic Science. The negative (ddH2O) and positive (9947A) controls were performed in our study following the manufacturer's instructions. The guidelines of *International Journal of Legal Medicine* and data uploaded recommendations of the International Society for Forensic Genetics (ISFG) were strictly followed in this study [3].

The distribution of allele frequencies and the statistical parameters, including power of discrimination (PD), power of exclusion (PE), polymorphism information content (PIC), paternity index (PI), cumulative paternity index (CPI), and the matching probability (MP), were calculated with the Modified-Powerstats software (Promega, Madison, WI, USA) [4]. Data of other 32 Chinese populations were referenced to conduct population comparisons. Hardy-Weinberg equilibrium (HWE) and Linkage disequilibrium (LD) were estimated using Arlequin version 3.5 [5]. The standard genetic distance between Chongqing Tujia and 32 Chinese populations was evaluated by PHYLIP 3.69 [6], and the results were applied to constructed the phylogenetic trees in Mega 7.0 [7]. The plot of PCA [8] and MDS were draw by MVSP 3.22 and SPSS 21, respectively.

Allele frequencies and parameters of the Plex system in Tujia were listed in Table S1. A total of 1012 alleles of this system were identified, and the allele frequencies ranged from 0.001 to 0.5761. The values of Ho, He, PD, and PE ranged from 0.1522 to 0.3913, 0.6087 to 0.8478, 0.7731 to 0.9651, and 0.3014 to 0.6906, respectively. The locus with the highest genetic polymorphism was D18S51, and the lowest was D1S1627. The PIC, TPI, and MP with range of 0.5340 to 0.8476, 1.2778 to 3.2857, and 0.0350 to 0.2270, respectively. The values of combined power of discrimination (CPD) and combined power of exclusion (CPE) in the new system were 0.99999999999999999999753 and 0.99999967, respectively. No significant difference were detected in Hardy-Weinberg equilibrium. The result of LD based on loci-by-loci comparisons of 23 autosomal STR loci showed that 16 pairs of loci present the p value less than 0.05 (Table S2). No association was noted in the analysis of LD after Bonferroni correction (0.05/506).

The genetic data of previously reported 32 Chinese populations (geographical location of the populations is presented in Fig. S1) were referenced to compare with our studied Tujia population. To explore genetic similarity and divergence among Chongqing Tujia and other 32 populations, three kinds of genetic distance were assessed firstly. In our present study, consistent results were found in Nei's genetic distance, Reynold's genetic distance, and Cavalli-Sforza genetic distance (Table S3), and graphical representation was presented in heatmap (Fig. S2). The smaller genetic distance was found between Chongqing Tujia and Hunn Han, Yugu, Hubei Tujia, and Sichuan Han. As the map shown, Chongqing is geographically bordered by Hunan, Sichuan, and Hubei, which can occur genetic communication or fusion between populations. Another interpretation is that populations originate from similar ancestors.

As MDS plots shown (Fig. S3), Han populations were gathered together which situated in the middle of plot, northern minorities were located in the upper quadrant, and southern populations were seated in the lower quadrant. The result of MDS lend support to the fact that Chongqing Tujia holds the similarity with Hubei Tujia and Sichuan Han. To synthetically analyzed genetic distance and relatedness between various populations, PCA was estimated based on the normalized allele frequencies. The findings (shown in Fig. S4) depicted that the first three components accounted for 49.77% of the total variance: the first, second, and third principal components interpreted 25.61%, 13.03%, and 11.13%, respectively. Three clear separations were revealed in the plot: the main Chinese Han populations were gathered together, two southern populations (Yunnan Bai and Yunnan Wa) formed the furthest part in the left of plot, and northern minorities located in the upper of plot. Chongqing Tujia population keeps the minimum difference with Hubei Tujia and Sichuan Han, which in conformity with the result of MDS.

As showed in the Fig. S5, the phylogenetic tree has two main branches: Yunnan Wa and Bai constituted one cluster, and the remaining ethnic groups formed another bigger branch in which show a significant departure between the Chinese Han and northern ethnic groups. Chongqing Tujia, Sichuan Han, and Hubei Tujia were gathered in a small branch, which was an agreement with the results of PCA and MDS. Results of population comparisons proved that the Chongqing Tujia has the most similar genetic structure to the Tujia in Hubei and the Han in Sichuan. Shu et al. [9] have studied populations relationship between Huann and 14 populations based on 17 Y-STR loci, and results manifested that Hunan Tujia and Han had no significant differences between that of Chongqing, indicating substantial homogeneity of two populations. Yang

et al. [10] confirmed that Chongqing Tujia keeps genetic similarity with the Southern Han populations based on 17 Y-STR loci, which was in accordance with our study.

The study here reported was undertaken to investigate the chromosomal polymorphism of new multiple system in Chongqing Tujia population. Our results provided evidence that these 23 autosomal STR loci could be effective for forensic application with its high polymorphism and efficacy parameter. In addition, the results will enrich the database of Chinese population, which could be conducive to anthropology, population origin, evolution, and other research in the future.

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Data accessibility The authors confirm that all the data in the research are available.

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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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