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POPULATION DATA

Genetic polymorphisms for 19 autosomal STR loci of Chongqing Han ethnicity and phylogenetic structure exploration among 28 Chinese populations

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Abstract The allele frequencies and forensic statistical parameters of 19 autosomal short tandem repeat (STR) loci (D8S1179, D21S11, D7S820, CSF1P0, D3S1358, THOl, D13S317, D16S539, D2S1338, D19S433, vWA, TPOX, D18S51, D5S818, FGA, D6S1043, Penta D, Penta E, and D12S391) included in the Goldeneye™ DNA ID system 20A kit were obtained in 671 Chinese Han individuals residing in Chongqing, Southwest China. All 19 STR loci were identified in agreement with the Hardy-Weinberg equilibrium. A total of 238 alleles were observed with corresponding allele frequencies that varied from 0.0007 to 0.5119. The combined power of discrimination and the combined probability of exclusion for 19 STR loci in the Chongqing Han population were 0.99999999999999999999998954 and 0.99999998387, respectively. The findings indicated that the 19 autosomal STR loci were highly polymorphic in the Chongqing Han population and can be used as a powerful tool in personal identification and parentage testing. Our genetic study enriched the Chinese local forensic reference database. Population comparisons and phylogenetic analyses revealed that genetic heterogeneity widely existed among the Chongqing Han, Xinjiang Uyghur, and Kazakh populations as well as demonstrated that genetic similarity was tightly associated with those of close geographic origin or of the same ethnic origin.

Keywords Genetic polymorphism · Short tandem repeat · Chongqing Han · Forensic genetics

Chongqing, being one of China's four direct-controlled municipalities, is situated in Southwest China. According to the 2010 demographic census, the population residing in Chongqing has exceeded 28 million people. The ethnic compositions in this area include Han Chinese, Tujia, Miao, and other ethnic groups. Han ethnicity, which is the largest ethnic group in the world, accounts for 93.6% of the population in Chongqing (https://en.wikipedia.org/wiki/Chongqing#cite_note-12).

Autosomal short tandem repeat (STR) loci have played a key role in personal identification and parentage testing. Recently, a larger number of genetic studies have concentrated on investigating the allele distributions and variations of autosomal STRs in Chinese nationalities. However, to the best of our knowledge, the population data and the genetic features of the Chongqing Han Chinese population remain unknown.

Our human genetic study protocol was approved by the Ethics Committee of Chongqing Medical University. Written informed consent from all of the study subjects was obtained before sample collection. A total of 671 (259 females and 412 males) EDTA anticoagulated peripheral blood samples were obtained from Han Chinese living in Chongqing, Southwest China (Fig. S1). Genomic DNA was extracted using the salting-out method [1], and a Nanodrop-1000 spectrophotometer (Thermo Scientific, Waltham, MA) was employed to measure the DNA concentration.

Xing Zou and YongGuo Li contributed equally to this study.

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Nineteen autosomal STR loci and a sex determination gene (Amelogenin) included in the Goldeneye™ 20A PCR amplification kit (Goldeneye Ltd., Beijing, China) were co-amplified using a GeneAmp PCR 9700 thermal cycler (Applied Biosystems, Foster City, CA, USA) according to the manufacturer's recommendations. The amplified products were segregated and detected using capillary electrophoresis and an ABI 3130 Genetic Analyzer (Applied Biosystems). Raw data analysis was performed using GeneMapper ID 3.2 software. Allele designations were determined by comparing the corresponding allelic ladders using the kit. Our experiments were performed at the Forensic Genetics Laboratory of the College of Basic Medicine, Chongqing Medical University, which was accredited by China National Accreditation Service for Conformity Assessment. The guidelines on population genetic data investigation recommended by the International Society for Forensic Genetics (ISFG) [2] and journal [3] were observed in the overall study.

Allele frequencies and corresponding forensic statistical parameters (the power of discrimination, PD; polymorphism information content, PIC; probability of exclusion, PE; and matching probability, MP) were calculated using modified PowerStats. Hardy-Weinberg equilibrium (HWE) and linkage disequilibrium were evaluated using Arlequin version 3.5 (http://cmpg.unibe.ch/software/arlequin35/). In our population comparisons and phylogenetic analyses, a total of 27 reference populations from different administrative or ethnic divisions were employed. The Fst and corresponding *p* values were computed using the Locus-by-Locus comparisons in Arlequin version 3.5.Nei's standard genetic distances among the aforementioned Chinese populations were computed using Phylip3.695 (http://evolution.genetics.washington.edu/phylip.html) and were deliberated by the neighbor joining tree in Mega 6.0 (http://www.megasoftware.net/).

A total of 671 Chinese Han individuals residing in Chongqing were genotyped in our study. All 19 STR loci included in the Goldeneye™ DNA ID system 20A kit were consistent with HWE except for D18S51 (0.0139), Penta D (0.0195), and D12S1338 (0.0182). No deviations from HWE were observed after the Bonferroni adjustment. As shown in Table S1, no evidence of linkage disequilibrium was found in the 19 STR loci. Allele frequencies and forensic statistical parameters regarding the 19 autosomal STR loci of the Chongqing Han population are presented in Table S2. A total of 238 alleles were identified with the corresponding allele frequencies varying from 0.0007 to 0.5119. The observed heterozygosity and

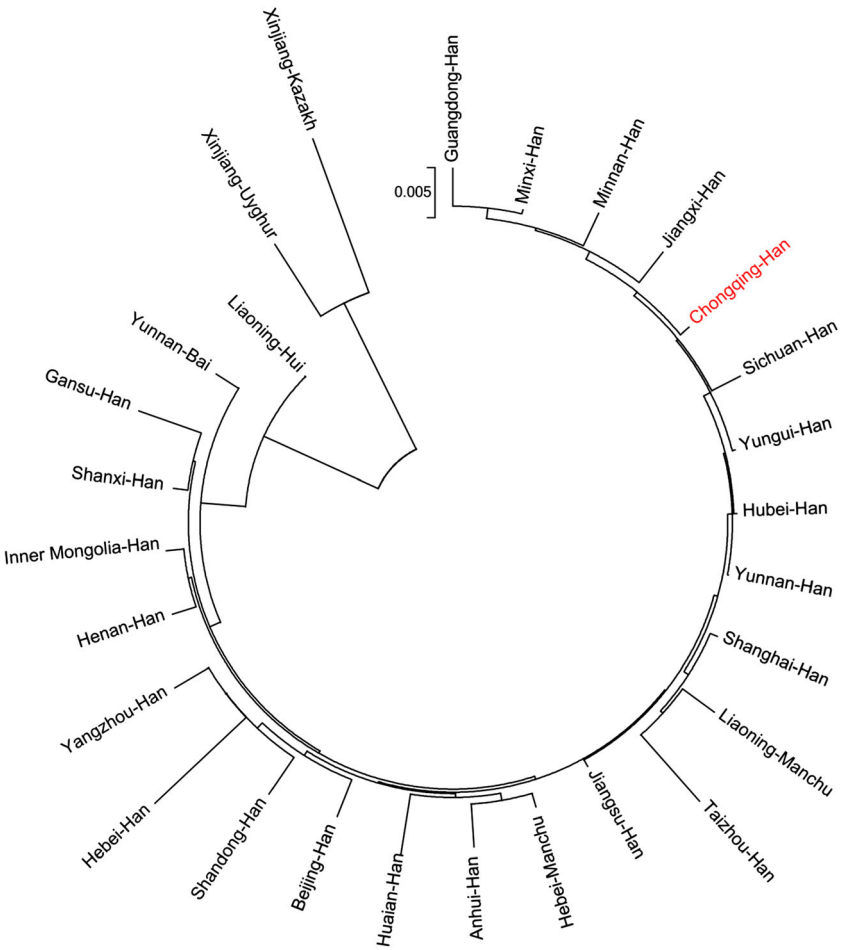

Fig. 1 The neighbor joining tree showing the phylogenetic relationship of the Chongqing Han population and 27 previously investigated Chinese populations. This tree was constructed based on 19 shared autosomal STR loci (D3S1358, D6S1043, D13S317, Penta E, D16S539, D18S51, D2S1338, CSF1PO, Penta D, TH01, vWA, D21S11, D7S820, D5S818, TPOX, D8S1179, D12S391, D19S433, FGA)

expected heterozygosity values spanned from 0.6110 (TPOX) to 0.9171 (Penta E) and from 0.6226 (TPOX) to 0.9171 (Penta E). The polymorphism information content, typical paternity index, and match probability were observed to range from 0.5579 (TPOX) to 0.9105 (Penta E), 1.2854 (TPOX) to 5.0833 (Penta E), and 0.0141 (Penta E) to 0.2061 (TPOX), respectively. The combined power of discrimination, the combined probability of exclusion as well as the match probability for 19 STR loci were 0.99999999999999999999998954, 0.99999998387, and 1.046×10^{-23}, respectively. These findings indicated that 19 autosomal STR loci are highly polymorphic and informative in the Chongqing Han population and can be used as a powerful tool in personal identification and parentage testing.

Population comparisons between the Chongqing Han population and one Uyghur [4], Kazakh [5], Bai [6], Hui [7], and two Manchu populations [8, 9] as well as 21 previous published Chinese populations distributed in different Chinese administrative divisions [10–27] were performed. The detailed information of geographic position and sample sizes of the included populations are shown in Fig. S1. The Fst and corresponding *p* value between the Chongqing Han population and the aforementioned populations in Locus-by-Locus comparisons are shown in Table S3. When the allele distributions for 19 STR loci of the Chongqing Han population were compared with other Chinese Han reference populations, no significant differences were observed with the exception of Guangdong Han at the locus of D2S1338, Hebei Han at the locus of D21S11, and Jiangxi Han at the locus of D19S433 after the Bonferroni Correction ($p < 0.0003$). When comparing the Chinese minority ethnic groups, significant differences were identified at four STR loci (D6S1043, D13S317, THO1, and vWA) with the Chinese Uyghur. In contrast, no significant difference was obtained with the minority ethnic groups of Bai and Manchu. The genetic similarity among several ethnic groups distributed in different administrative divisions or different ethnic groups may be affected by random marriage, cultural background, and human migration. At the same time, genetic heterogeneity significantly existed between the Chinese Uyghur and Han populations.

To explore the genetic diversity and phylogenetic characteristics among Chinese populations, the genetic distance was calculated and presented in Table S4. The short genetic distances were observed between the Chongqing Han and Yungui Han (0.0023) and Yunnan Han (0.0028). However, large genetic distances were found between the Chongqing Han and Xinjiang Kazakh (0.0572), followed by Chongqing Han and Xinjiang Uyghur (0.0487). Subsequently, as shown in Fig.1, phylogenetic relationships among 28 Chinese populations are visualized in a neighbor joining tree. All 28 Chinese populations were clustered in two groups: one group consisted of two populations residing in Xinjiang (the Uyghurs and Kazakhs); the other comprised 26 populations clustered together. The Chongqing Han population first clustered with the geographic close populations (Jiangxi, Sichuan, and Yungui Han). The results demonstrated that genetic affinity was associated with geographic distance and ethnic group origin.

The allele frequencies and forensic statistical parameters of 19 STR loci included in the Goldeneye™ DNA ID system 20A kit for 671 Chinese Han individuals residing in Chongqing were studied. A total of 238 alleles were identified and the corresponding allele frequencies varied from 0.0007 to 0.5119. The combined power of discrimination and the combined probability of exclusion for 19 STR loci were 0.99999999999999999999998954 and 0.99999998387, respectively. These findings indicated that 19 autosomal STR loci are highly polymorphic and informative in the Chongqing Han population and can be used as a powerful tool in personal identification and parentage testing. Our genetic study enriched the Chinese local forensic reference database. Population comparisons and phylogenetic analyses not only revealed that genetic heterogeneity widely exists between the Chongqing Han and Xinjiang Uyghur and Kazakh but also demonstrated that genetic similarity was associated with those of close geographic or ethnic origin.

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Compliance with ethical standards

Conflicts of interest The authors declare that they have no potential conflicts of interest to disclose.

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