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Genetic investigation of Chinese she ethnic based on autosomal STRs and X-STRs

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ABSTRACT

The She ethnic is a large minority in China with approximately 700,000 individuals. For more than one thousand years, they mainly resided in Zhejiang and Fujian provinces. To obtain a better understanding of the genetic background of She, we investigate 21 autosomal STRs (A-STRs) and 16 X-STRs in 296 unrelated healthy individuals from Zhejiang She population. Allele frequencies and forensic parameters prove these markers are useful for forensic application. We also investigate the genetic background with the two types of markers. Nei genetic distances between She and Eastern Han population are always the lowest, regardless of the markers used for analysis. Although the tested STRs are located on different chromosomes with different inheritance laws, A-STRs and X-STRs provided in general congruent phylogenetic signal and similar cluster among compared groups. These results demonstrated that geographic isolation and interactions play significant roles in differentiation of genetic constitution of ethnic groups.

1. Introduction

Autosomal STR (A-STR) markers have been used to study genetic polymorphism for a long time. Over the past 30 years, a number of studies have been performed in different populations worldwide with A-STRs [1]. X-STRs are emerging popular genetic markers for genetic investigation in forensic community. In this study, we try to explore the genetic background of She ethnic minority with the two types of markers. She, an ancient population in China, mostly clustered in small, relatively isolated villages in mountainous regions. This isolation has helped preserve the integrity of their language and traditions.

2. Materials and methods

Blood samples were collected from 296 unrelated individuals (129 males and 167 females) from the Zhejiang She population with written informed consents. QIAamp DNA Blood Mini Kit (QIAGEN, Hilden, Germany) was used to extract Genomic DNA. All samples were amplified with Goldeneye™ DNA ID 22NC Kit and Goldeneye™ DNA ID 17X Kit in the GeneAmp PCR System 9700 (Thermo Fisher Scientific, USA). Goldeneye™ DNA ID 22NC Kit contains 17 non−CODIS loci (D19S253, D6S477, D22GATA198B05, D15S659, D8S1132, D3S3045, D14S608, D17S1290, D3S1744, D2S411, D18S535, D13S325, D7S1517, D10S1435, D11S2368, D4S2366, D7S3048) and 3 CODIS loci (D3S1358, D1S1656 and D10S1248), while Goldeneye™ DNA ID 17X Kit contains 16 X-STRs (DXS6795, DXS9902, DXS8378, HPRTB, GATA165B12, DXS7132, DXS7424, DXS6807, DXS6803, GATA172D05, DXS6800, DXS10134, GATA31E08, DXS6810, DXS6789, DXS10159). The PCR products were separated by capillary electrophoresis using POP-4 polymers on a Genetic Analyzer ABI 3130xl (Thermo Fisher Scientific, USA) and the data were analyzed with GeneMapper ID Analysis Software. The A-STRs dataset has successfully passed STRidER quality control (STRidER dataset reference STR000177) [2].

Arlequin v3.5 was utilized to calculate Hardy-Weinberg equilibrium (HWE) and to perform linkage disequilibrium (LD) test. The allele frequencies and forensic parameters of 21 A-STRs were calculated using PowerStat V1.2 (Promega, Madison, WI). Forensic parameters of 16 X-STRs, including polymorphism information content (PIC), heterozygosity (HET), power of discrimination in males (PD_M) and females (PD_F), and mean paternity exclusion chance in duos (MEC_D) and trios (MEC_T), were estimated based on an online tool provided by Forensic ChrX-STR database (http://www.chrx-str.org). The F_{st} pairwise genetic

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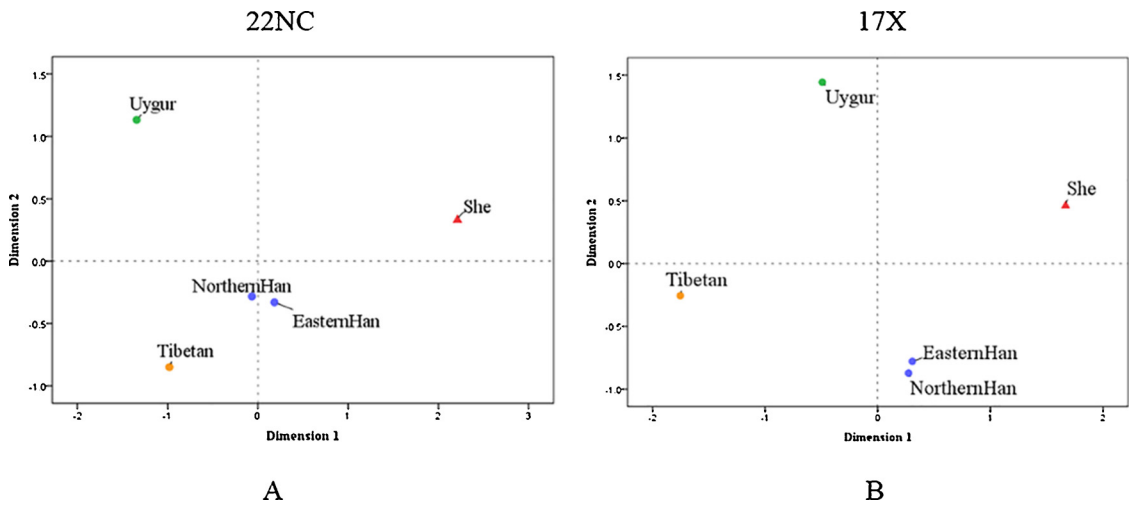

Fig. 1. MDS plots based on F_{st} values between She population (marked with red triangle) and other four different Chinese populations (For interpretation of the references to colour in this figure legend, the reader is referred to the web version of this article).

distances were explored by Arlequin V3.5. Multidimensional scaling (MDS) plots were generated based on F_{st} values using the SPSS Statistics 24 software (SPSS, Chicago, IL).

3. Results and discussion

For the 21 A-STRs, all loci follow the HWE after Bonferroni's correction; the 21 loci are highly informative with HET ranging from 0.699 (D2S441) to 0.892 (D11S2368), PD from 0.871 (D3S1358) to 0.968 (D7S1517) and PIC from 0.67 (D3S1358) to 0.86 (D7S3048). Since the 21 loci are independent from each other after LD testing, the combined power of discrimination (CPD) and combined power of exclusion (CPE) are 1–6.4E-25 and 0.999999996, respectively. The allele frequencies of 16 X-STRs in males and females show no significant difference at any locus ($p > 0.05$), therefore, pooled frequencies of males and females were used to calculate forensic parameters. The 16 X-STRs follow HWE in females. The HET range from 0.3244 (DXS6800) to 0.8680 (DXS10134). The discrimination power in females (PD_F) and males (PD_M) are 0.999999997 and 0.999999999999993, respectively. The mean exclusion chance in duos (MEC_D) and trios (MEC_T) are 0.9999931 and 0.99999997, respectively. Above results demonstrate that both the 21 A-STRs and 16 X-STRs can provide highly polymorphic information for forensic identification and paternity testing.

To further explore the genetic relationship among populations, we compared A-STRs and X-STRs data of She with previously published data from Eastern Han [3,4], Northern Han [1,5], Tibetan [6] and Uygur [7] on the same 21 A-STRs and 11 overlapping X-STRs. The F_{st} value between She and Eastern Han are always the lowest, regardless of the A-STRs or X-STRs were used ($F_{st} = 0.0249$ with A-STRs and $F_{st} = 0.0054$ with X-STRs). Based on the F_{st} genetic distance, the MDS plots generated by A-STRs and X-STRs are listed as Fig.1A and Fig.1B, respectively. In Fig.1, the She population is closely related to Eastern Han and distantly related to Tibetan, which is consistent with the historical and geographical background of the compared populations. Similar results were obtained in the previous studied based on 12 X-STRs [8] or 30 InDels [9]. However, the observed genetic relationships could be complicated by many factors including the origins of samples, the choice of genetic markers and coverage of reference populations. Thus, great care must be taken when relating one ethnic group to another, as there may have been intermixture with now formally extinct populations that contributes to current population genetic structure.

4. Conclusion

The forensic parameters indicate that the investigated 21 A-STRs and the 16 X-STRs are highly polymorphic and informative in the She population. These non−CODIS STRs can be useful in paternity cases with mutations or complex cases, while the X-STRs show higher efficiency in special kinship investigations involving mainly female offspring [10]. Although X-STRs tell a different story from A-STRs, they provided in general congruent phylogenetic signal and similar cluster among the five ethnic groups, which demonstrated that geographic isolation and interactions play significant roles in differentiation of genetic constitution of ethnic groups.

Declaration of Competing Interest

None.

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