

Genetic polymorphism of 30 autosomal InDel loci in Chinese Hainan Li population

Ziwei Ye, Jing Liu, Hong Zhu, Zheng Wang, Yiping Hou*

Institute of Forensic Medicine, West China School of Basic Science and Forensic Medicine, Sichuan University, Chengdu, 610041, China

ARTICLE INFO

Keywords:

Insertion/Deletion polymorphism
Population genetics
Hainan Li population

ABSTRACT

A novel genetic marker, Insertion/Deletion polymorphism (InDel) shows remarkable potential for forensic DNA applications. Hainan Island is the southernmost and the second largest island in China, of which the Li ethnic group is regarded as the original inhabitants. In this study, 207 individual samples of Li ethnic group from Hainan were genotyped using Investigator DIPplex kit which contains 30 autosomal InDels and Amelogenin. Allele frequency and forensic parameters were calculated for these loci. Several deviations from Hardy-Weinberg equilibrium (HWE) and linkage disequilibrium (LD) may indicate founder effect in the Li ethnic group. The combined power of discrimination (CPD) and the cumulative probability of exclusion (CPE) reached 0.9999999992912 and 0.9861, respectively. These results suggested that the kit was effective for personal identification in Hainan Li population. The population comparisons through the Nei's standard genetic distance (R_{st}), phylogenetic tree, multidimensional scaling analysis (MDS), principal component analysis (PCA), and STRUCTURE analyses along continental divisions manifested that the 30 InDels panel had a certain inter-continental differentiation ability.

1. Introduction

A novel short-length diallelic polymorphism, Insertion and deletion polymorphism (InDel), is regarded as an extraordinary promising genetic marker combining the advantages of both STR and SNP analysis and could play a certain role in forensic identification, ancestry inference and population genetic analysis [1]. The Qiagen Investigator DIPplex Kit containing Amelogenin and 30 InDels have been widely testified among multiple populations to verify its forensic and biogeographic efficacy. Hainan Island, the second largest island in China, is located at the southernmost tip of China. It's widely believed that the Li population are the original inhabitants of Hainan Island as the descendants of the ancient Yue tribes [2]. According to the 2010 National Population Census, the Hainan Li group has more than one million population accounting for 15% of the whole population on the island [3]. In the present study, we analyzed the genetic diversity of 30 autosomal InDels in Hainan Li group and validated the efficacy of the Investigator DIPplex Kit in this population.

2. Materials and methods

A total of 207 blood samples were collected from unrelated individuals from Li ethnic group living in Hainan Island with written

informed consent. Human genomic DNA was isolated by PureLink Genomic DNA Mini Kit (Thermo Fisher Scientific), quantified by the Nanodrop-2000C (Thermo Fisher Scientific), amplified by the Investigator DIPplex commercial kit (Qiagen), genotyped by capillary electrophoresis on Applied Biosystems 3130 Genetic Analyzer (Thermo Fisher Scientific) and then analyzed by GeneMapper ID-X version 1.5 software (Thermo Fisher Scientific). Allele frequencies and forensic parameters, including observed heterozygosity (H_o), expected heterozygosity (H_e), matching probability (PM), power of discrimination (PD), probability of exclusion (PE) and typical paternity index (TPI), as well as the Hardy-Weinberg equilibrium (HWE) and linkage disequilibrium (LD) tests were analyzed using the STRAF online software. To inspect the genetic divergence between Hainan Li and other populations, a series of population comparison analyses were performed. Genotype data of other 22 populations was obtained from previous studies (Asian [4,5], European [6], American [7], African [8]) Genotype data of other 22 populations was obtained from previous studies. The R_{st} value was calculated based on allele frequencies using the Phylip3.695 package. The phylogenetic tree based on the neighbor-joining (N-J) method was constructed in the Molecular Evolutionary Genetics Analysis (MEGA v7.0) software. The MDS was carried out based on R_{st} using the SPSS software v19.0 (IBM SPSS). The principal component analysis (PCA) scatter plot was depicted based on allele

* Corresponding author at: 3-17 Renmin South Road, Chengdu, 610041, China.
E-mail address: forensic@scu.edu.cn (Y. Hou).

<https://doi.org/10.1016/j.fsigss.2019.09.042>

Received 26 August 2019; Accepted 23 September 2019

Available online 05 October 2019

1875-1768/ © 2019 Elsevier B.V. All rights reserved.

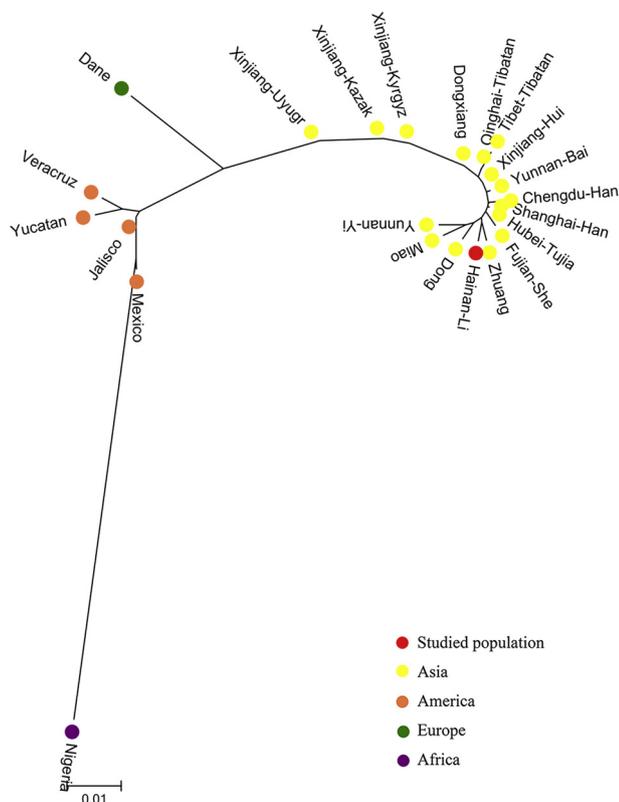


Fig. 1. Phylogenetic tree among 23 populations.

frequencies by the MVSP v3.22 software. The structure analysis based on genotype data of the 30 autosomal InDels was performed with the STRUCTURE v.2.3.4 software.

3. Results and discussion

No remarkable deviation from the HWE was observed from any InDel loci with a 95% significance level except HLD40, HLD83 and HLD93, probably due to random sampling. It's worth noting that the H_o sometimes exceeds the H_e in population with a small founding population size or which has experienced bottleneck events [9]. In this case, for HLD83 and HLD90, H_o were observed much higher than H_e , which may indicated a founder effect [10] regarding the special history of the Li ethnic group. According to a previous research, founder effect was also identified in the Li Ethnic group in Hainan Island by phylogenetic analysis of both Y-STR and Y-SNP [3]. A few significant LD were observed in a total of 435 interclass correlation tests with the significance level of 95% (data not shown), also probably due to a strong founder effect [11]. The insertion allele frequencies among the 30 InDel loci were in the range of 0.0459 (HLD39) to 0.9251 (HLD118). The H_o values varied from 0.0725 (HLD39) to 0.5942 (HLD93), and the H_e values ranged from 0.0878 (HLD39) to 0.5012 (HLD92). The lowest PM was 0.3577 (HLD88), whereas the highest PM was discovered at HLD39 (PM = 0.8478). The PD, PE, and TPI values ranged from 0.2435 (HLD118) to 0.6423 (HLD88), from 0.0046 (HLD39) to 0.2840 (HLD93), and from 0.5391 (HLD39) to 1.2321 (HLD93), respectively. The CPD and the CPE were 0.99999999292 and 0.9861, respectively. These results suggested that the 30 InDels panel was effective for personal identification but may not powerful enough to perform paternity

test in Hainan Li population. The R_{st} values between 23 populations showed that Li had the closest relationship with Dong ($R_{st} = 0.0080$) and kept the largest R_{st} (0.2059) with the African Nigeria. Moreover, the result of phylogenetic tree reconstruction (Fig. 1), MDS, PCA, STRUCTURE analysis were consistent with each other, all conformed to the geographical distribution. The Hainan Li population had genetic affinities with most Asian groups and possessed the largest difference from the African group.

4. Conclusion

In conclusion, the population data on the 30 InDels indicated that the Investigator DIPplex commercial kit could be a promising tool for personal identification and a complementary tool for kinship testing in Hainan Li population and had a certain intercontinental differentiation ability. For our investigated populations, the Hainan Li had genetic affinities with Most Asian groups and kept the largest genetic difference from the African group. Furthermore, the deviation from HWE and LD of several locus may indicate a founder effect in Hainan Li population. To better dissect the background and origin of Hainan Li groups, more ancestry informative InDels and SNPs should be selected and validated in the future.

Declaration of Competing Interest

The authors declare that they have no conflicts of interest.

Acknowledgment

This study was supported by grant from National Key R&D Program of China (2016YFC0800703) and from the National Natural Science Foundation of China (81571854) and the Open project of Shanghai Key Laboratory of Forensic Medicine (Academy of Forensic Science) (KF1814).

References

- [1] J.L. Weber, D. David, J. Heil, et al., Human diallelic insertion/deletion polymorphisms, *Am. J. Hum. Genet.* 71 (2002) 854–862.
- [2] M.S. Peng, J.D. He, H.X. Liu, et al., Tracing the legacy of the early Hainan Islanders—a perspective from mitochondrial DNA, *BMC Evol. Biol.* 11 (2011) 46.
- [3] M. Song, Z. Wang, Y. Zhang, et al., Forensic characteristics and phylogenetic analysis of both Y-STR and Y-SNP in the Li and Han ethnic groups from Hainan Island of China, *Forensic Sci. Int. Genet.* 39 (2019) e14–e20.
- [4] Y.L. Wei, C.J. Qin, H. Dong, et al., A validation study of a multiplex INDEL assay for forensic use in four Chinese populations, *Forensic Sci. Int. Genet.* 9 (2014) e22–25.
- [5] T. Mei, C.M. Shen, Y.S. Liu, et al., Population genetic structure analysis and forensic evaluation of Xinjiang Uigur ethnic group on genomic deletion and insertion polymorphisms, *Springerplus* 5 (2016) 1087.
- [6] S.L. Friis, C. Borsting, E. Rockenbauer, et al., Typing of 30 insertion/deletions in Danes using the first commercial indel kit—Mentype® DIPplex, *Forensic Sci. Int. Genet.* 6 (2012) e72–74.
- [7] G. Martinez-Cortes, M. Garcia-Aceves, A.F. Favela-Mendoza, et al., Forensic parameters of the Investigator DIPplex kit (Qiagen) in six Mexican populations, *Int. J. Legal Med.* 130 (2016) 683–685.
- [8] G. Hefke, S. Davison, M.E. D'Amato, Forensic performance of Investigator DIPplex indels genotyping kit in native, immigrant, and admixed populations in South Africa, *Electrophoresis* 36 (2015) 3018–3025.
- [9] J. Eun Kim, R.E. Withler, C. Ritland, et al., Genetic variation within and between domesticated chinook salmon, *Oncorhynchus tshawytscha*, strains and their progenitor populations, *Environ. Biol. Fishes* 69 (2004) 371–378.
- [10] D. Kaczmarczyk, M. Luczynski, P. Brzuzan, Genetic variation in three paddlefish (*Polyodon spathula* Walbaum) stocks based on microsatellite DNA analysis, *Czech J. Anim. Sci.* 57 (2012) 345–352.
- [11] W.G. Hill, Estimation of effective population size from data on linkage disequilibrium, *Genet. Res. (Camb)* 38 (1981) 209–216.