Genetic analysis of 17 Y-STR loci in Han population from Shandong Province in East China

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Dear Editor,

Shandong Province is located in East China, including two sub-provincial and fifteen prefecture-level cities (Fig. S1). According to the 2009 census, the population of Shandong reached 95,793,065, and Han made up the largest ethnic group.

In the present study, a total of 734 unrelated male Han Chinese individuals were collected from Shandong Province. These samples were collected from twelve sub-provincial or prefecture-level cities, including Jinan (n=59), Binzhou (n=30), Dezhou (n=114), Dongying (n=76), Heze (n=29), Jining (n=41), Linyi (n=122), Rizhao (n=29), Tai'an (n=21), Weifang (n=21), Yantai (n=122) and Zibo (n=70). Peripheral blood samples were collected from these individuals after acquiring their informed consent. Genomic DNA was extracted using the phenol-chloroform method. A total of 17 Y-STRs were analyzed, including DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, YGATAH4 and DYS385a/b. Polymerase chain reaction (PCR) was performed using the AmpFISTR YfilerTM PCR Amplification kit (Thermo Fisher Scientific Company, Carlsbad, USA) in the GeneAmp PCR System 9700 (Thermo Fisher Scientific Company) according to the manufacturer's instruction. PCR products were separated by capillary electrophoresis in ABI PRISM 3130xL Genetic Analyzer (Thermo Fisher Scientific Company). The GeneMapper ID software

53 v3.2 (Thermo Fisher Scientific Company) was used for genotype assignment. DNA
54 typing and assignment of nomenclature were based on the ISFG recommendations
55 [1,2].

56 We estimated pairwise Rst between twelve Han populations of Shandong using
57 Arlequin v3.5 (10,000 permutations)[3]. In this analysis, DYS385 a/b was excluded,
58 and DYS389B was obtained by subtracting the allele of DYS389I from that of
59 DYS389II. The haplotype frequency was estimated using direct counting method.
60 Haplotype diversity was estimated as described by Nei[4]: $HD = (n/n - 1)(1 - \sum p_i^2)$,
61 where n is the sample size and p_i is the frequency of the i th haplotype. Moreover,
62 pairwise Rst was estimated between Shandong Han and other Chinese populations in
63 previous reports using Arlequin v3.5[3], and the results were visualized by
64 multidimensional scaling plot (MDS) using R v3.1.2 (<http://www.r-project.org>).

65 We observed significant difference between Dezhou and Linyi after Bonferroni
66 correction ($p = 0.05/66 = 0.00075758$, 66 are the number of the test). Significant
67 differences were also observed between Zibo and the other six Shandong Han
68 populations including Binzhou, Dezhou, Jinan, Linyi, Rizhao and Yantai (Table S1).
69 The results showed that the haplotype frequency of Zibo was slightly different from
70 other Han populations in Shandong of the present study, and this may require further
71 evaluation on larger dataset.

72 A total of 701 different haplotypes were observed among all individuals of
73 Shandong Han population, of which 676 were unique, 21 were shared by two
74 individuals, 3 were shared by three individuals and 1 was shared by seven individuals

75 (Table S2). The haplotype diversity reached 0.99981.

76 We estimated pairwise Rst between the Han population of Shandong in this study
77 and other reports involving Chinese populations. A total of fourteen Chinese Han
78 [5-13] and twelve ethnic populations [6, 14-21] were obtained from earlier reports.
79 Among them, two Han populations were sampled from Shandong Province [7, 13].
80 Overall, 8,056 reference samples were analyzed. Figure S2 showed that Shandong
81 Han is genetically close to other Han populations and to Manchu, Mongolian, and
82 Xibe. As shown in Table S3, Shandong Han of the present study had no significant
83 difference between Shandong, China [Han], whereas showed significant difference
84 between Jining, China [Han] after Bonferroni correction ($p = 0.05/351 =$
85 0.00014245, 351 are the number of the test). In addition, we observed that Shandong
86 Han of the present study showed significant differences between the other five
87 Chinese Han populations (Jiangsu, China [Han], Zhejiang, China [Han], Luzhou,
88 China [Han], Shanxi, China [Han] and Henan, China [Han]). Significant differences
89 were also observed between Shandong Han and nearly all Chinese ethnic populations
90 except Manchu and Xibe.

91 In conclusion, we report 17 Y-STR loci's haplotype frequencies and forensic
92 parameters of the Han population in Shandong Province. We believe the data is
93 valuable for both forensics and population genetics.

94 Our data has been submitted to YHRD, and received the accession number:
95 Shandong, China [Han], n=734, YA004081.

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99 The study follows the new guidelines for publication of population data requested
100 by the journal[22, 23].

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