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Short Communication

Molecular phylogenetic study of the Iranians based on polymorphic markers

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ABSTRACT

The application of polymorphic markers in construction of phylogenetic trees has been documented. Five polymorphic markers located in the *PAH* gene region including PAH-*Bgl*II, PAH-*Pvu*II(A), PAH-*Eco*RI, PAH-*Msp*I and PAH-STR were selected for analysis of phylogenetic relationships of the Iranians with 15 other populations of the world. The lowest genetic distance was observed between the Iranians and populations residing in Adygei (an ethnic group of the Russian Caucasus), Russia and Druze (a Middle Eastern group). However, East Asian populations including Han, Japanese and Cambodians, Khmer or the Oceanians (Melanesian, Nasioi) showed high genetic distance with the Iranians. The data suggested that the Iranians might have relatively close evolutionary history with the populations residing in Russia rather than East Asian populations. This study provided the first new molecular insight into the evolutionary history of the Iranian population.

1. Introduction

The origin and history of human populations across the world are considerably attractive topics in population genetics. The allele frequency variation at polymorphic markers has proven to be useful for studying the human evolution and migration (Agrawal and Khan, 2005; Katsuyama et al., 1998). During the last few decades, microsatellite markers and single nucleotide polymorphisms (SNPs) have been utilized for the survey of relationship between human populations. It has been shown that these polymorphic markers were informative in constructing evolutionary trees and elucidating questions concerning the human evolution (Agrawal and Khan, 2005; Ayub et al., 2003; Bowcock et al., 1994; Deka et al., 1995; Goldstein and Pollock, 1997; Jorde et al., 1997; Katsuyama et al., 1998).

The mutation rate of single nucleotide polymorphisms (SNPs) was estimated at approximately 2.3×10^{-8} per base per generation (Nachman and Crowell, 2000). According to the low mutation rate of these markers, it seems unlikely that these markers could have reoccurred during the evolution of humans. Therefore, the study of SNPs could facilitate investigation into recent human history. Moreover, microsatellite markers or short tandem repeats (STRs) evolve at a faster rate than SNPs and are located throughout the human genome. The mutation rate varies among STR markers. The average mutation rate for STR markers was estimated to about 10^{-4} to 10^{-3} per base per generation, making these markers ideal for phylogenetic analysis of populations (Crawford, 2007; Jobling et al., 2004). Among the polymorphic markers, those located on autosomal chromosomes have some advantages over the markers on Y chromosome and mitochondrial DNA (mtDNA). The Y chromosome and mtDNA inherited from paternal and maternal sides, respectively. Moreover, the autosomal polymorphic markers transmitted bi-parentally and could provide more information about the history of both sexes (Crawford, 2007).

To date no reports are available on the phylogenetic relationships of the Iranians with other world populations by use of autosomal polymorphic markers. In the present study, we have used allele frequency data of five polymorphic markers from the Iranian population to evaluate the genetic relationship between the Iranians and 15 other populations of the world.

2. Materials and methods

2.1. Polymorphic markers and population data

The genotyping data from 250 healthy unrelated Iranian individuals, which were identified in our previous studies, were included in the present study (Fazeli and Vallian, 2009a, 2009b). The data for 15 other world populations including Biaka, Mbuti, Yoruba, Druze (a middle Eastern group), Adygei, Russian, Han, Japanese, Cambodians, Melanesian, Yakut, Mexican, Yucatan, Karitiana and Surui were taken from the ALFRED (Allele Frequency Database) website (Cheung et al., 2000,

Abbreviations: ALFRED, Allele Frequency Database; DNA, deoxyribonucleic acid; dNTP, deoxyribonucleoside triphosphate; mtDNA, mitochondrial DNA; NJ, Neighbor Joining; PAGE, PA-gel electrophoresis; PAH, phenylketonuria; RFLP, restriction-fragment length polymorphism; SNPs, single nucleotide polymorphisms; STR, short tandem repeat.

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Table 1
D_A genetic distances (×100) between the studied populations based on allele frequency data obtained from five polymorphic markers located in the *PAH* gene region.

Geographic region	Population	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16
Asia	1. Iranian	5.1	7.4	3.6	1.9	1.1	1.4	6.3	6.7	8.9	4.5	3.5	12.0	6.4	6.5	8.5
Africa	2. Biaka		3.6	2.1	7.3	4.1	5.1	10.6	11.6	11.3	11.3	7.5	4.1	1.0	7.2	3.3
Africa	3. Mbuti			7.4	11.1	6.6	9.1	6.3	7.4	9.6	13.1	7.6	9.3	3.7	8.2	5.2
Africa	4. Yoruba				5.5	3.1	3.2	11.4	12.2	10.9	9.9	6.7	5.4	2.5	7.6	*6.0*
Europe	5. Druze					1.1	1.1	6.5	6.3	9.0	3.1	2.2	13.7	8.3	7.6	8.2
Europe	6. Adygei						0.8	4.9	5.3	8.3	4.0	1.5	9.0	4.3	4.6	5.1
Europe	7. Russian							7.2	7.3	8.6	3.2	2.6	11.4	6.2	6.1	7.3
East Asia	8. Han								0.3	4.2	8.3	3.1	15.8	10.3	12.0	9.7
East Asia	9. Japanese									4.5	7.3	2.9	17.0	11.5	13.0	9.8
East Asia	10. Cambodians										11.9	7.0	15.2	11.8	17.7	13.7
Oceania	11. Melanesian											3.3	19.1	11.7	7.9	9.8
Siberia	12. Yakut												12.7	6.6	5.7	5.5
North America	13. Mexican													3.1	11.5	5.4
North America	14. Yucatan														5.3	2.1
South America	15. Karitiana															5.7
South America	16. Surui															

Rajeevan et al., 2005). The polymorphic markers included were PAH-*Bgl*II, PAH-*Pvu*II(A), PAH-*Eco*RI, PAH-*Msp*I and PAH-STR (Dworniczak, et al., 1991; Goltsov, et al., 1993; Wedemeyer, et al., 1991).

2.2. Statistical and phylogenetic assessment

The allele frequency of the markers in the Iranian population was estimated using the GENEPOP website (Raymond and Rousset, 1995). The population relationship was investigated through genetic distance measurements based on the allele frequency of the studied polymorphic markers. Genetic distance between the Iranians and the other populations was calculated by the modified Cavalli-Sforza distance (D_A). The phylogenetic trees were constructed by the Neighbor Joining (NJ) method (Saitou and Nei, 1987). The advantage of this method for construction of phylogenetic trees over the other methods such as the UPGMA (Sokal and Sneath, 1963) has been documented (Livshits and Nei, 1990; Takezaki and Nei 1996). The calculation of genetic distance and construction of phylogenetic trees were carried out by use of the POPTREE2 computer program (Takezaki et al., 2010). The confidence of phylogenetic trees was assessed by bootstrapping method. The bootstrap test was performed with 1000 replications.

3. Results

In this study, the allele frequency data of five polymorphic markers were used to evaluate the genetic relationships of the Iranians with other populations. A total of 15 different populations were compared with the Iranian population, and the genetic distance between the studied populations was estimated by using the allelic frequency of the markers. These populations included Biaka, Mbuti, Yoruba, Druze, Adygei, Russian, Han, Japanese, Cambodians, Melanesian, Yakut, Mexican, Yucatan, Karitiana and Surui. The geographical region of these populations is shown in Table 1. The genetic distance obtained from these polymorphic markers is also presented in Table 1. As presented in Fig. 1, the obtained

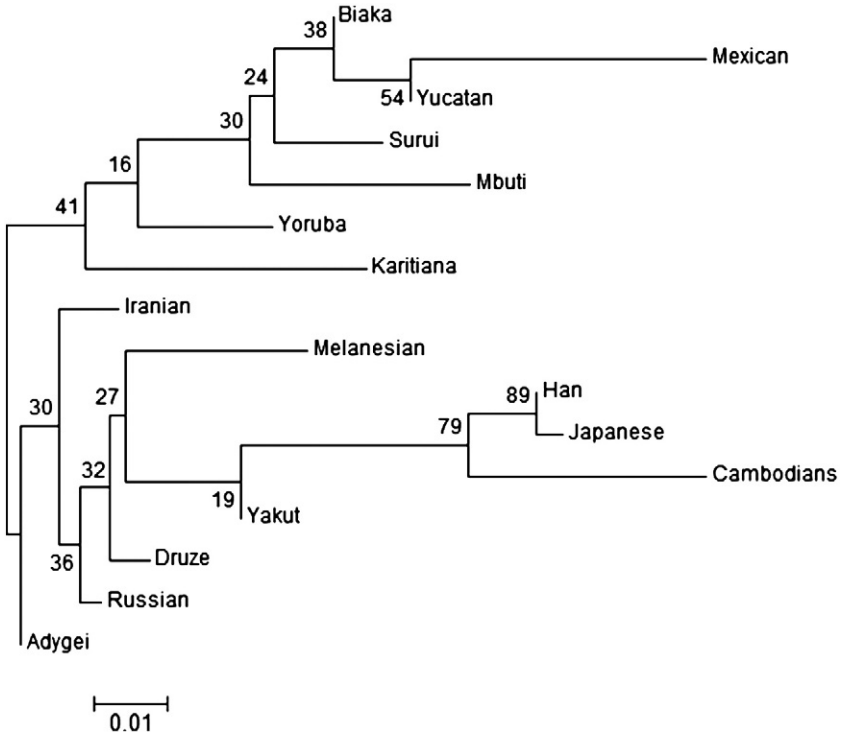

Fig. 1. Neighbor joining tree of 16 populations along with bootstrap support value. The trees were constructed by using D_A distances in Table 1.