Available online at www.sciencedirect.com
ScienceDirect
Forensic Science International: Genetics 2 (2008) e9–e10
www.elsevier.com/locate/fsig

Announcement of population data

Genetic polymorphisms of 16 Y chromosomal STR loci in Korean population

Soon Hee Kim [a,c], Nam Ye Kim [a], Seung Beom Hong [a], Nam Soo Cho [b], Jong Jin Kim [a], Myun Soo Han [a], Won Kim [c,*]

[a] DNA Analysis Division, National Institute of Scientific Investigation, Seoul 158-097, Republic of Korea
[b] Department of Forensic Medicine, Central District Office, National Institute of Scientific Investigation, Daejeon 305-348, Republic of Korea
[c] School of Biological Sciences, Seoul National University, San56-1, Sillim-dong, Gwanak-gu, Seoul 151-742, Republic of Korea

Received 22 May 2007; received in revised form 30 October 2007; accepted 3 November 2007

Abstract

Allele frequencies and haplotypes of 16 Y chromosomal STR loci included in the AmpFlSTR® Yfiler™ system were obtained from a sample of 526 unrelated Korean male individuals. A total of 478 haplotypes were observed in the 526 individuals studied, of which 440 were unique. The overall haplotype diversity for the 16 Y-STR loci was 0.9996, and the discrimination capacity was 0.9087. We found 13 atypical alleles, including null, duplicated and microvariant alleles. Microvariants have been characterized by sequencing, 14.1 allele at DYS458 showing the flanking site mutation, 13.1 and 15.2 allele at DYS385a/b showing changes in the repeat structure.

Keywords: Y chromosomal STRs; Haplotype; Korean; Population; Microvariant

Population: Buccal swab and blood samples were obtained from 526 unrelated healthy Korean male individuals living in all the major province of South Korea, i.e., Seoul-Gyeonggi, Chungcheong, Jeolla, Gyeongsang, Gangwon and Jeju. All the samples were collected with their informed consent. Each of these samples was amplified using the Powerplex® 16 (Promega) and the AmpFlSTR® Identifiler™ kit (Applied Biosystems) and shown to be unique.

Extraction: DNA was isolated from buccal swab and blood samples by the DNA IQ™ system (Promega).

PCR: The 16 markers of DYS456, DYS389I, DYS390, DYS389II, DYS458, DYS19, DYS385a/b, DYS393, DYS391, DYS439, DYS635, DYS392, YGATA H4, DYS437, DYS438 and DYS448 were co-amplified using the AmpFlSTR® Yfiler™ kit (Applied Biosystems) following the recommendations.

Typing: Amplified PCR products were subjected to capillary electrophoresis on a 3730 Genetic Analyzer (Applied Biosystems) according to the manufacture's recommendation.

Quality control: Use of the control DNA and allelic ladders provided by manufacturer of the kit.

Sequence analysis: For DYS458, the primers designed for sequence analysis were as follows: primer 1, 5′-GGG TGG TGG AGG TTA CTG TG-3′; primer 2, 5′-GGA TGG TCT CGA TTT CCT GA-3′. For sequencing of locus DYS385a/b, the following primers were used: primer 1, 5′-AGC ATG GGT GAC AGA GCT A-3′; primer 2, 5′-TGG GAT GCT AGG TAA AGC TG-3′. Amplicons of DYS385a/b were cloned using the TOPO TA Cloning kit (Invitrogen, Germany). Sequencing was performed using the BigDye Terminator Cycle Sequencing kit (Applied Biosystems).

Access to the data: Available upon request: soondi@nisi.go.kr

Data analysis: Haplotypes and allele frequencies were estimated by gene counting. Haplotypes and gene diversities were estimated according to Nei [1]. Haplotype discrimination capacity (DC) was calculated as DC = h/n, where h is the total number of different haplotypes, and n is the total number of individuals in the sample. AMOVA analysis and calculation of F_{ST} values were performed using Arlequin 3.11 [2]. The bilocal system DYS385a/b was not considered.

Results: See Tables 1–3 in Supplementary materials.

* Corresponding author. Tel.: +82 2 880 6695; fax: +82 2 872 1993.
E-mail address: wonkim@plaza.snu.ac.kr (W. Kim).

1872-4973/$ – see front matter © 2007 Elsevier Ireland Ltd. All rights reserved.
doi:10.1016/j.fsigen.2007.11.001

Other remarks: A total of 478 different haplotypes were detected from 526 Korean male individuals, of which 440 haplotypes were unique. Thirty, six and two haplotypes were shared by two, three and four individuals, respectively (Supplementary Table 1). Overall haplotype diversity in the 16 Y-STR loci was 0.9996, and a discrimination capacity was 0.9087. These values are higher than the values for the Southern and the Central population of Korea [3,4]. These results indicate a high potential for differentiating between male individuals. Gene diversity values of these loci ranged from 0.3169 (DYS391) to 0.9603 (DYS385a/b) (Supplementary Table 2).

Supplementary Table 3 summarized the haplotypes with rare allelic variants, including null, duplicated and microvariant alleles, which were not reported previous of Korean population [3–5]. These allelic variants were reproducible and confirmed by re-amplification using both the Yfiler™ kit and the PowerPlex® Y (Promega). Y-STR null alleles were detected at DYS448 in six individuals. Null alleles were also observed at the DYS19 and DYS393. Duplicated alleles were found at the following single copy Y-STR markers: DYS390 and DYS458. A microvariant allele, typed as 14.1 was observed for DYS458. DNA sequence analysis revealed the insertion of base 'A' at base 1 downstream from the 14 repeats of a GAAA motif. This was named as 14(D1Ains) according to Gusmão et al. [6]. Two microvariant alleles, typed as 13.1 and 15.2 were observed for DYS385a/b. DNA sequence analysis revealed that microvariants were due to an irregular repeat unit in the STR repeat region. Full repeat sequences of 13.1 and 15.2 were $[GAAA]_{10}A[GAAA]_3$ and $[GAAA]_{11}GA[GAAA]_4$, respectively.

Our present haplotype data were compared with previous published Asian populations [7–9]. Seventeen haplotypes were shared between Korean and Japanese. The most common haplotypes were Ht274, Ht277, Ht319 and Ht322, which were shared by five individuals. We searched these four haplotypes in the YHRD database (http://www.yhrd.org) among 5544 haplotypes in a set of 40 East Asian populations. Ht277 (DYS19-DYS389I-DYS389II-DYS390-DYS391-DYS392-DYS393-YS385a/b-DYS438-DYS439 15-14-30-22-10-13-13-10/20-13-12) showed the most frequent shared haplotype, 32 matches were found in the YHRD database, i.e. 31 in Japanese, 1 in Sino Tibetan.

A comparative analysis showed that the Korean population has significant differences from Japanese population (F_{ST} = 0.00123, P = 0.00000) [7], Chinese Han population (F_{ST} = 0.00074, P = 0.00391) [8], Malays in Malaysia (F_{ST} = 0.00075, P = 0.00000), Chinese in Malaysia (F_{ST} = 0.00103, P = 0.00000), and Indians in Malaysia (F_{ST} = 0.00079, P = 0.00000) [9]. Population pairwise comparisons using both the studied haplotypes and PowerPlex® Y haplotypes [4] showed the same result in the context of the relationships of neighboring populations with the Korean-Chinese Han groups being the most related genetically. There was no significant difference between the studied population and other populations from Korea, showing P = 0.45946 with [3], P = 0.11712 with [4], and P = 0.56757 with [5].

This paper follows the guidelines for publication of population data requested by the journal [6,10].

Acknowledgment

This work was supported by a grant from the Korean Ministry of Science and Technology (M10640020002-06N4002-00210).

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.fsigen.2007.11.001.

References

[1] M. Nei, Molecular Evolutionary Genetics, Columbia University Press, New York, 1987, pp. 176–179.
[2] L. Excoffier, G. Laval, S. Schneider, Arlequin ver. 3.0: an integrated software package for population genetics data analysis, Evol. Bioinformatics Online 1 (2005) 47–50.
[3] B.W. Chun, S.C. Shin, Y.J. Kim, K.S. Kim, D.H. Choi, K.H. Kim, J.Y. Kim, H.S. Kang, Allele frequencies and haplotypes of the STR loci of the PowerPlex Y-system in southern populations from Korea, Forensic Sci. Int. 148 (2005) 225–231.
[4] J.H. Hwang, H. Lee, Y.A. Lee, M.S. Han, W. Kim, I. Jin, N.S. Cho, Haplotypes for 12 Y-chromosomal STR loci in a Korean population (the central region), Forensic Sci. Int. 168 (2007) 73–84.
[5] M.J. Park, H.Y. Lee, J.E. Yoo, U. Chung, S.Y. Lee, K.J. Shin, Forensic evaluation and haplotypes of 19 Y-chromosomal STR loci in Koreans, Forensic Sci. Int. 152 (2005) 133–147.
[6] L. Gusmão, J.M. Butler, A. Carracedo, P. Gill, M. Kayser, W.R. Mayr, N. Morling, M. Prinz, L. Roewer, C. Tyler-Smith, P.M. Schneider, DNA commission of the International Society of Forensic Genetics (ISFG): an update of the recommendations on the use of Y-STRs in forensic analysis, Forensic Sci. Int. 157 (2006) 187–197.
[7] N. Mizuno, H. Nakahara, K. Sekiguchi, K. Yoshida, M. Nakano, K. Kasai, 16 Y chromosomal STR haplotypes in Japanese, Forensic Sci. Int. 174 (2008) 71–76.
[8] J. Yan, H. Tang, Y. Liu, Y. Jing, Z. Jiao, Q. Zhang, J. Gao, L. Shang, H. Guo, J. Yu, Genetic polymorphisms of 17 Y-STRs haplotypes in Chinese Han population residing in Shandong province of China, Leg. Med. 9 (2007) 196–202.
[9] Y.M. Chang, R. Perumal, P.Y. Keat, D.L.C. Kuehn, Haplotype diversity of 16 Y-chromosomal STRs in three main ethnic populations (Malays, Chinese and Indians) in Malaysia, Forensic Sci. Int. 167 (2007) 70–76.
[10] P. Lincoln, A. Carracedo, Publication of population data of human polymorphisms, Forensic Sci. Int. 110 (2000) 3–5.