Jian Tie, ${ }^{1}$ M.D.; Yuka Suzuki, ${ }^{1}$ B.Sc.; Etsuko Iwakami, ${ }^{1}$ B.Sc.; Shigemi Oshida, ${ }^{1}$ M.D.; and George Sensabaugh, ${ }^{2}$ Ph.D.

## The Polymorphisms of Four Y-Chromosome Short Tandem Repeat Loci in Chinese and Japanese Populations


#### Abstract

POPULATION: A total of 155 unrelated healthy Chinese males living in Shenyang (Liaoning province) and 186 unrelated Japanese healthy males living in Tokyo.


KEYWORDS: forensic science, DNA typing, Y-chromosome, short tandem repeat, population genetics

The DNA was isolated from blood by the standard phenol/chloroform method and precipitated with ethanol. The PCR was carried out using a total reaction volume of $25 \mu \mathrm{l}$ with the following conditions: an initial denaturation step at $95^{\circ} \mathrm{C}$ for 5 min followed by 30 cycles of denaturation at $94^{\circ} \mathrm{C}$ for 60 s , primer annealing at $60^{\circ} \mathrm{C}$ for 45 s and primer extension at $72^{\circ} \mathrm{C}$ for 60 s , with a final extension at $60^{\circ} \mathrm{C}$ for 40 min . For separating the PCR products of the multiplex in one gel, we redesigned the primers for GATA A4 ( $5^{\prime}$-TCT CGA GTT GTT ATG GTT TTA GGT C- $3^{\prime}$ and GCC TGG CTT GGA ATT CTT TT-3') and GATA A10 ( $5^{\prime}$-TCA TCC ATC CTC TTT CTT TCT CTC C- $3^{\prime}$ and TGG AGA TAG TGG GTG GAT TG-3') to allele sizes $205 \sim 229$ and $104 \sim 128 \mathrm{bp}$. The primer sequences of GATA A7.2 and GATA C4 loci were according to White et al. (1). The PCR products were separated on $6.5 \%$ denaturing polyacrylamide gel ( $5 \% \mathrm{C}$ ) contains 7 M urea. The bands of allele fragments were visualized by silver staining. DNA typing was performed using allelic ladders that were constructed using the corresponding allele fragments identified by sequence analysis (ABI PRISM ${ }^{\text {TM }} 310$ Genetic Analyzer).
The observed allele frequencies are shown in Table 1. The most diverse loci were GATA C4, GATA A4 and GATA A10 in Chinese, and GATA A10 and GATA C4 in Japanese. Gene diversity was estimated according to Nei (2). Basically, higher gene diversity values were found in Chinese than in the Japanese at each locus. The allele frequency distributions at the four loci were not significantly different between the two populations (A4, A10 and $\mathrm{C} 4, p<0.95$; A7.2, $p<0.90$ ). These four Y-chromosome STR loci were combined to

[^0]TABLE 1-Allele frequency and gene diversity of four Y-STR loci in Chinese (Ch) and Japanese (Jp) population.

| Allele | Y-A4 |  | Y-A7.2 |  | Y-A10 |  | Y-C4 |  |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: |
|  | Ch | Jp | Ch | Jp | Ch | Jp | Ch | Jp |
| 7 |  |  |  | 0.013 |  |  |  |  |
| 8 |  | 0.004 | 0.006 | 0.278 |  |  |  |  |
| 9 |  |  | 0.142 | 0.616 | 0.006 |  |  |  |
| 10 | 0.071 | 0.022 | 0.561 | 0.068 | 0.006 | 0.018 | 0.168 | 0.012 |
| 11 | 0.258 | 0.177 | 0.239 | 0.017 | 0.045 | 0.066 | 0.271 | 0.327 |
| 12 | 0.432 | 0.608 | 0.052 | 0.008 | 0.329 | 0.442 | 0.219 | 0.482 |
| 13 | 0.213 | 0.185 |  |  | 0.413 | 0.381 | 0.168 | 0.102 |
| 14 | 0.026 | 0.004 |  |  | 0.150 | 0.058 | 0.090 | 0.041 |
| 15 |  |  |  |  | 0.045 | 0.035 | 0.078 | 0.029 |
| 16 |  |  |  |  | 0.006 |  | 0.006 | 0.008 |
| Gd* | 0.700 | 0.567 | 0.609 | 0.541 | 0.700 | 0.654 | 0.813 | 0.651 |

* gene diversity.
produce a total of 147 different haplotypes (93 in Chinese and 72 in Japanese; Table 2).

All the alleles identified increased in regularly by 4 base pair increments. Sequencing results showed that the major repeat structure was simple GATA. The GATA C4 locus was only (TCTA) 4 (TGTA) $)_{2}$ $(T C T A)_{2}$ (TGTA)n was observed in both populations. However, a different sequence structure $(\text { TCTA })_{4}(\text { TGTA })_{2}(T C T A)_{2}(T G T A)_{1}$ (TCTA) ${ }_{11}$ was found in two Japanese individuals. The common repeat structure of GATA C4 was (GATA) ${ }_{n}$ based on White et al., it is allele 11. But Gonzālez-Neira et al. call it allele 20 (3). The PCR amplification efficiency for GATA A4 locus was poor using White's primers (3), but this problem was not happed by the newly designed primer in this study.

TABLE 2-Haplotype (H) of four Y-STR loci in Chinese (Ch) and Japanese (Jp) population.

| H | A4 | Y-STR |  | C4 | n |  | H | A4 | Y-STR |  | C4 | n |  |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: |
|  |  | A7.2 | A10 |  | $\overline{\mathrm{Ch}}$ | Jp |  |  | A7.2 | A10 |  | $\overline{\mathrm{Ch}}$ | Jp |
| H1 | 9 | 9 | 13 | 15 |  | 1 | H75 | 12 | 9 | 14 | 10 | 1 |  |
| H2 | 10 | 9 | 11 | 12 |  | 1 | H76 | 12 | 9 | 14 | 11 | 1 | 18 |
| H3 | 10 | 9 | 12 | 13 | 1 |  | H77 | 12 | 9 | 14 | 12 |  | 3 |
| H4 | 10 | 9 | 12 | 14 | 1 |  | H78 | 12 | 9 | 15 | 10 | 1 |  |
| H5 | 10 | 9 | 13 | 13 | 1 |  | H79 | 12 | 9 | 15 | 11 |  | 2 |
| H6 | 10 | 9 | 13 | 14 | 1 |  | H80 | 12 | 9 | 15 | 12 |  | 1 |
| H7 | 10 | 10 | 12 | 11 | 2 |  | H81 | 12 | 10 | 10 | 11 | 1 |  |
| H8 | 10 | 10 | 12 | 12 | 2 | 1 | H82 | 12 | 10 | 11 | 11 |  | 1 |
| H9 | 10 | 10 | 13 | 11 | 1 |  | H83 | 12 | 10 | 11 | 12 |  | 1 |
| H10 | 10 | 10 | 13 | 13 | 1 |  | H84 | 12 | 10 | 11 | 14 | 1 |  |
| H11 | 10 | 10 | 14 | 14 | 1 |  | H85 | 12 | 10 | 12 | 10 |  |  |
| H12 | 11 | 8 | 11 | 12 |  | , | H86 | 12 | 10 | 12 | 11 | 3 | 1 |
| H13 | 11 | 8 | 12 | 12 |  | 6 | H87 | 12 | 10 | 12 | 12 | 5 | 1 |
| H14 | 11 | 8 | 13 | 11 |  | 1 | H88 | 12 | 10 | 12 | 13 | 3 |  |
| H15 | 11 | 8 | 13 | 12 |  | 1 | H89 | 12 | 10 | 12 | 14 | 3 |  |
| H16 | 11 | 9 | 11 | 11 | 1 |  | H90 | 12 | 10 | 13 | 10 | 3 |  |
| H17 | 11 | 9 | 11 | 12 | 1 |  | H91 | 12 | 10 | 13 | 11 | 6 |  |
| H18 | 11 | 9 | 12 | 11 |  | 2 | H92 | 12 | 10 | 13 | 12 | 1 | 2 |
| H19 | 11 | 9 | 12 | 12 |  | 2 | H93 | 12 | 10 | 13 | 13 |  | 2 |
| H20 | 11 | 9 | 12 | 13 |  | 2 | H94 | 12 | 10 | 14 | 10 | 3 |  |
| H21 | 11 | 9 | 12 | 14 | 1 | 2 | H95 | 12 | 10 | 14 | 11 | 4 |  |
| H22 | 11 | 9 | 12 | 15 | 2 |  | H96 | 12 | 10 | 14 | 12 | 1 |  |
| H23 | 11 | 9 | 13 | 11 | 1 | 1 | H97 | 12 | 10 | 14 | 13 | , |  |
| H24 | 11 | 9 | 13 | 13 |  | 1 | H98 | 12 | 10 | 15 | 12 | 1 |  |
| H25 | 11 | 9 | 13 | 15 | 1 |  | H99 | 12 | 11 | 9 | 12 |  |  |
| H26 | 11 | 9 | 14 | 11 |  | 2 | H100 | 12 | 11 | 12 | 12 | 1 |  |
| H27 | 11 | 10 | 11 | 11 |  | 1 | H101 | 12 | 11 | 12 | 13 | 2 |  |
| H28 | 11 | 10 | 11 | 13 | 1 |  | H102 | 12 | 11 | 13 | 10 | 2 |  |
| H29 | 11 | 10 | 11 | 15 |  | , | H103 | 12 | 11 | 13 | 12 | 2 |  |
| H30 | 11 | 10 | 12 | 11 | 3 | 1 | H104 | 12 | 11 | 13 | 13 | 4 |  |
| H31 | 11 | 10 | 12 | 12 | 3 |  | H105 | 12 | 11 | 13 | 15 | 2 |  |
| H32 | 11 | 10 | 12 | 13 | 2 | 1 | H106 | 12 | 11 | 14 | 11 | 1 |  |
| H33 | 11 | 10 | 12 | 14 | 2 |  | H107 | 12 | 11 | 15 | 10 | 1 |  |
| H34 | 11 | 10 | 13 | 10 | 3 |  | H108 | 12 | 11 | 15 | 12 | 1 |  |
| H35 | 11 | 10 | 13 | 11 | 4 |  | H109 | 12 | 12 | 12 | 12 |  | 1 |
| H36 | 11 | 10 | 13 | 12 | 2 | 5 | H110 | 12 | 12 | 13 | 10 | 1 |  |
| H37 | 11 | 10 | 13 | 15 | 3 |  | H111 | 12 | 12 | 13 | 12 | 2 |  |
| H38 | 11 | 11 | 12 | 12 | 2 |  | H112 | 12 | 12 | 15 | 11 |  | 1 |
| H39 | 11 | 11 | 12 | 13 | 1 |  | H113 | 12 | 12 | 16 | 11 | 1 |  |
| H40 | 11 | 11 | 12 | 14 | 1 |  | H114 | 13 | 8 | 11 | 12 |  | 1 |
| H41 | 11 | 11 | 13 | 11 | 3 |  | H115 | 13 | 8 | 12 | 12 |  | 7 |
| H42 | 11 | 11 | 14 | 15 | 1 |  | H116 | 13 | 8 | 12 | 13 |  | 1 |
| H43 | 11 | 11 | 15 | 15 | 1 |  | H117 | 13 | 8 | 15 | 12 |  | 1 |
| H44 | 11 | 12 | 13 | 10 | 1 |  | H118 | 13 | 9 | 11 | 11 | 1 |  |
| H45 | 12 | 8 | 11 | 14 |  | 1 | H119 | 13 | 9 | 11 | 12 |  | 1 |
| H46 | 12 | 8 | 12 | 11 |  | 1 | H120 | 13 | 9 | 12 | 11 |  | 3 |
| H47 | 12 | 8 | 12 | 12 |  | 13 | H121 | 13 | 9 | 12 | 12 |  | 6 |
| H48 | 12 | 8 | 12 | 14 |  | 1 | H122 | 13 | 9 | 12 | 13 | 1 | 2 |
| H49 | 12 | 8 | 12 | 15 |  | 1 | H123 | 13 | 9 | 13 | 11 |  | 5 |
| H50 | 12 | 8 | 13 | 11 |  | 1 | H124 | 13 | 9 | 13 | 12 |  | 5 |
| H51 | 12 | 8 | 13 | 12 |  | 6 | H125 | 13 | 9 | 14 | 11 |  | 1 |
| H52 | 12 | 8 | 13 | 13 |  | 2 | H126 | 13 | 10 | 12 | 12 | 2 |  |
| H53 | 12 | 8 | 13 | 14 |  | 2 | H127 | 13 | 10 | 12 | 13 | 3 |  |
| H54 | 12 | 8 | 13 | 15 | 1 |  | H128 | 13 | 10 | 13 | 11 | 2 | 1 |
| H55 | 12 | 8 | 14 | 11 |  | 3 | H129 | 13 | 10 | 13 | 13 | 2 |  |
| H56 | 12 | 8 | 14 | 12 |  | 1 | H130 | 13 | 10 | 13 | 16 | 1 |  |
| H57 | 12 | 8 | 14 | 15 |  | 1 | H131 | 13 | 10 | 14 | 10 | 3 | 2 |
| H58 | 12 | 8 | 15 | 12 |  | 1 | H132 | 13 | 10 | 14 | 12 | 2 |  |
| H59 | 12 | 9 | 10 | 11 |  | 1 | H133 | 13 | 10 | 15 | 10 | 1 |  |
| H60 | 12 | 9 | 10 | 12 |  | 2 | H134 | 13 | 10 | 15 | 11 | 1 |  |
| H61 | 12 | 9 | 11 | 11 | 1 | 1 | H135 | 13 | 11 | 11 | 11 | 1 |  |
| H62 | 12 | 9 | 11 | 12 |  | 1 | H136 | 13 | 11 | 13 | 10 | 1 |  |
| H63 | 12 | 9 | 11 | 13 |  | 1 | H137 | 13 | 11 | 13 | 11 | 2 |  |
| H64 | 12 | 9 | 12 | 11 |  | 2 | H138 | 13 | 11 | 13 | 12 | 2 |  |
| H65 | 12 | 9 | 12 | 12 |  | 15 | H139 | 13 | 11 | 13 | 13 | 1 |  |
| H66 | 12 | 9 | 12 | 13 | 1 | 4 | H140 | 13 | 11 | 13 | 14 | 1 |  |
| H67 | 12 | 9 | 12 | 14 | 1 | 1 | H141 | 13 | 11 | 14 | 10 | , |  |
| H68 | 12 | 9 | 12 | 15 | 1 |  | H142 | 13 | 11 | 14 | 11 | 2 |  |
| H69 | 12 | 9 | 13 | 11 |  | 9 | H143 | 13 | 12 | 12 | 12 | 1 |  |
| H70 | 12 | 9 | 13 | 12 | 1 | 7 | H144 | 13 | 12 | 13 | 10 | 2 |  |
| H71 | 12 | 9 | 13 | 13 |  | 2 | H145 | 14 | 10 | 13 | 13 | 1 |  |
| H72 | 12 | 9 | 13 | 14 | 1 |  | H146 | 14 | 10 | 14 | 10 | 1 |  |
| H73 | 12 | 9 | 13 | 15 |  | 1 | H147 | 14 | 11 | 13 | 10 | 1 |  |
| H74 | 12 | 9 | 13 | 16 |  | 1 |  |  |  |  |  |  |  |

Haplotype diversity value was 0.992 for Chinese and 0.970 for Japanese.

The complete data set is available to any interested researcher at http://www.med.nihon-u.ac.jp/~legalmed/jtie/Y-STR.doc.

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## [PubMed]

Additional information and reprint requests:
Jian Tie, M.D.
Department of Legal Medicine
Nihon University School of Medicine
30-1, Oyaguchi-kamimachi
Itabashi-ku, Tokyo 173-8610
Japan


[^0]:    ${ }^{1}$ Department of Legal Medicine, Nihon University School of Medicine, Tokyo, Japan.
    ${ }^{2}$ Forensic and biomedical science, Division of Public Health, University of California, Berkeley, CA.

