

FOR THE RECORD

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Analysis of Y-STR Loci in a Population Sample from Northeast China

POPULATION: A total of 141 unrelated Chinese Han male individuals living in Liaoning in northeast China.

KEYWORDS: forensic science, short tandem repeat, Y-chromosome, haplotype, Han population, Liaoning of northeast China, DNA typing, population genetics, DYS389I, DYS389II, DYS439, DYS438, DYS392, DYS393, DYS19, DYS390, DYS391, DYS385

The distribution of allele frequencies and haplotypes for 10 microsatellites (short tandem repeat (STR)) loci, DYS389I, DYS389II, DYS439, DYS438, DYS392, DYS393, DYS19, DYS390, DYS391, and DYS385 on the Y-chromosome were determined in 141 DNA samples of unrelated Chinese Han male individuals living in Liaoning of northeast China, using Y-PLEXTM 5 and Y-PLEXTM 6 (ReliaGene Technologies, New Orleans, LA). The Y-STR genotypes were determined using the ABI PRISMTM 310 Genetic Analyzer and the GeneScan analysis software (Applied Biosystems, Foster City, CA). During the 10 Y-STR loci analysis, altogether 65 alleles and 135 haplotypes were identified in these Chinese male individuals, and the haplotype diversity was raised to 0.9998. The results indicate that DYS389I, DYS389II, DYS439, DYS438, DYS392, DYS393, DYS19, DYS390, DYS391, and DYS385 are useful Y-specific STR markers for forensic sciences.

DNA extraction: The DNA was extracted from the blood of 141 unrelated healthy Chinese Han male individuals living in Liaoning by using John's method (1).

PCR amplification: For amplification of 10 Y-STR loci, a multiplex polymerase chain reaction (PCR) was performed using a combination of the Y-PLEXTM 5 and Y-PLEXTM 6 kit (ReliaGene Technologies). The PCR amplification reactions were performed in a GeneAmp PCR system 9700 (Applied Biosystems) under the following conditions: 30 cycles of initial denaturation at 95°C for 10 min, amplification with denaturation at 94°C for 30 s, annealing at 59°C for 1 min, and extension at 70°C for 1 min, followed by a final extension at 60°C for 60 min.

Genotyping: Detection of the amplified PCR products for 10 Y-STR loci was carried out using the ABI PRISMTM 310 Genetic Analyzer (Applied Biosystems). 0.5 µL of PCR products were mixed with 20 µL of formamide and 1 µL of GS-500 ROX (Applied Biosystems) and denatured for 5 min. Allele designations

were performed using the GeneScan analysis software (Applied Biosystems) and reference-sequenced ladders (Y-PLEXTM 5 and Y-PLEXTM 6, ReliaGene Technologies).

Data analysis: Analysis of data was carried out using the ARLEQUIN package (2). Gene diversity and haplotype diversity were estimated according to Nei (3).

Results: See Tables 1 and 2.

The distribution of allele frequencies and gene diversity for the 10 Y-STR loci were estimated from 141 Chinese Han male individuals living in Liaoning in northeast China. A total of 66 alleles were identified in DYS389I, DYS389II, DYS439, DYS438, DYS392, DYS393, DYS19, DYS390, DYS391, and DYS385. The gene diversity values for each locus ranged from 0.4869 (DYS391) to 0.8921 (DYS385a/b). A total of 135 haplotypes were observed in 141 northeast Chinese male individuals for haplotypes constructs of the 10 Y-STR loci; the haplotype diversity was raised to 0.9998.

Other remarks: The comparison of allele frequencies and haplotype diversity between the 141 Chinese samples and other Chinese samples (4,5) showed no special variation for 10 Y-STR loci. Some micro alleles (11 & 15 of DYS389I, 14 of DYS439, 10 of DYS392) were observed in this study, and allele 6 of DYS391 and 7 of DYS392 were not found. The distribution of allele frequencies between different populations showed considerable differences in 10 Y-STR loci (6–10). The gene diversity of each locus and 10 loci haplotype diversity were compared with Japanese, Italian, Caucasian, African American, Hispanic, Pakistani, and Bangladeshi populations (Table 3). Gene diversity values between different populations are different for the 10 Y-STR loci. The haplotype diversity values of 10 Y-STR loci were high in all populations.

These gene and haplotype diversities indicated that the 10 Y-STR polymorphism markers are highly discriminating in the Chinese population, and they may be very powerful for establishing Y-STR database, understanding human origin, paternity testing, and personal identification.

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

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TABLE 1—Allele frequencies of 10 Y-STR loci in 141 Chinese Han male individuals of Liaoning, China.

Allele	DYS389I	DYS389II	DYS439	DYS438	DYS392	DYS393	DYS19	DYS390	DYS391	DYS385a/b
8	—	—	—	—	—	—	—	—	0.0071	—
9	—	—	—	0.0213	—	—	—	—	0.0709	0.0036
10	—	—	0.0709	0.6454	0.0071	0.0071	—	—	0.6879	0.0461
11	0.0071	—	0.2553	0.2128	0.2553	0.0213	—	—	0.1915	0.1312
12	0.4681	—	0.5532	0.0567	0.1277	0.5319	—	—	0.0355	0.1950
13	0.2766	—	0.1135	0.0638	0.2908	0.2411	0.0497	—	0.0071	0.1702
14	0.2411	—	0.0071	—	0.2766	0.1135	0.2624	—	—	0.0709
15	0.0071	—	—	—	0.0425	0.0851	0.4326	—	—	0.0532
16	—	—	—	—	—	—	0.1915	—	—	0.0461
17	—	—	—	—	—	—	0.0638	—	—	0.0745
18	—	—	—	—	—	—	—	—	—	0.0532
19	—	—	—	—	—	—	—	—	—	0.0780
20	—	—	—	—	—	—	—	—	—	0.0567
21	—	—	—	—	—	—	—	0.0213	—	0.0142
22	—	—	—	—	—	—	—	0.0638	—	0.0071
23	—	—	—	—	—	—	—	0.3972	—	—
24	—	—	—	—	—	—	—	0.3191	—	—
25	—	—	—	—	—	—	—	0.1844	—	—
26	—	0.0071	—	—	—	—	—	0.0142	—	—
27	—	0.1064	—	—	—	—	—	—	—	—
28	—	0.3050	—	—	—	—	—	—	—	—
29	—	0.2553	—	—	—	—	—	—	—	—
30	—	0.2198	—	—	—	—	—	—	—	—
31	—	0.0851	—	—	—	—	—	—	—	—
32	—	0.0142	—	—	—	—	—	—	—	—
33	—	0.0071	—	—	—	—	—	—	—	—
GD	0.6508	0.7801	0.6152	0.5342	0.7609	0.6429	0.7058	0.7068	0.4869	0.8921

STR, short tandem repeat; GD, gene diversity.

TABLE 2—The observed haplotypes for 10 Y-STR loci in 141 Chinese Han male individuals living in Liaoning, China.

Haplotype	DYS389I	DYS389II	DYS439	DYS438	DYS392	DYS393	DYS19	DYS390	DYS391	DYS385a/b	No.
H 1	11	28	12	10	13	12	14	24	11	13/19	1
H 2	12	26	11	10	11	13	15	23	10	12/13	1
H 3	12	27	11	10	11	11	14	23	11	11/11	1
H 4	12	27	11	10	14	12	14	21	10	11/13	1
H 5	12	27	12	10	13	12	15	23	10	10/19	1
H 6	12	27	12	10	12	14	16	22	10	13/18	1
H 7	12	27	12	10	12	13	16	23	11	14/15	1
H 8	12	27	12	11	12	15	15	25	10	12/18	1
H 9	12	27	12	11	14	10	15	23	10	12/16	1
H 10	12	27	12	11	14	12	17	24	10	13/20	1
H 11	12	28	13	10	14	12	16	25	10	13/13	1
H 12	12	28	10	10	13	12	14	25	10	12/19	1
H 13	12	28	11	9	13	12	15	24	10	12/20	1
H 14	12	28	11	9	13	12	15	23	11	13/17	1
H 15	12	28	11	11	14	12	14	22	10	10/19	1
H 16	12	28	11	10	11	15	15	23	10	12/18	1
H 17	12	28	11	10	12	13	15	24	10	11/11	1
H 18	12	28	11	10	11	12	16	23	10	10/10	1
H 19	12	28	11	10	12	15	14	23	10	11/17	1
H 20	12	28	11	10	14	12	15	23	10	9/11	1
H 21	12	28	11	10	14	12	16	24	10	13/22	1
H 22	12	28	11	10	11	12	17	24	10	12/20	1
H 23	12	28	11	10	13	12	14	23	11	19/21	1
H 24	12	28	12	11	15	14	15	24	9	14/16	1
H 25	12	28	12	11	14	15	16	23	10	11/15	1
H 26	12	28	12	12	14	13	16	23	10	10/12	1
H 27	12	28	12	10	13	12	15	23	10	12/13	2
H 28	12	28	12	10	13	13	13	23	10	13/17	1
H 29	12	28	12	10	11	12	14	21	10	14/16	1
H 30	12	28	12	10	11	12	16	24	13	12/17	1
H 31	12	28	12	11	11	12	17	24	11	12/19	1
H 32	12	28	12	11	13	14	14	26	10	14/19	1
H 33	12	28	12	11	12	12	15	24	12	14/20	1
H 34	12	28	12	10	14	12	16	25	12	14/17	1
H 35	12	28	13	10	12	13	17	25	11	17/20	1
H 36	12	28	13	10	15	13	15	23	10	12/18	1
H 37	12	28	14	10	11	12	14	23	10	11/19	1

TABLE 2—Continued.

Haplotype	DYS389I	DYS389II	DYS439	DYS438	DYS392	DYS393	DYS19	DYS390	DYS391	DYS385a/b	No.
H 38	12	29	12	10	11	12	14	24	10	11/12	2
H 39	12	29	12	10	15	13	15	23	10	15/20	1
H 40	12	29	12	10	13	12	15	24	9	12/19	1
H 41	12	29	12	10	11	14	15	23	10	11/18	1
H 42	12	29	12	10	13	12	16	24	10	12/15	1
H 43	12	29	11	11	13	13	16	21	10	12/19	1
H 44	12	29	12	10	12	12	17	23	10	14/14	1
H 45	12	29	12	11	13	12	13	25	10	13/15	1
H 46	12	29	13	13	13	12	15	23	12	10/17	1
H 47	12	29	11	10	14	12	15	24	10	10/10	1
H 48	12	29	10	10	11	14	16	24	10	12/20	1
H 49	12	29	11	9	11	13	15	23	11	13/14	1
H 50	12	29	12	13	14	13	15	23	10	11/11	1
H 51	12	29	12	10	11	12	15	23	11	12/12	1
H 52	12	29	12	10	14	12	14	24	10	13/19	2
H 53	12	30	10	10	15	13	14	24	11	11/14	1
H 54	12	30	11	10	13	13	13	23	10	17/19	1
H 55	12	30	12	11	13	12	16	25	9	11/13	1
H 56	12	30	12	11	11	13	14	23	10	13/13	1
H 57	12	30	10	10	14	13	14	23	11	11/18	1
H 58	12	30	11	13	14	14	16	25	11	15/19	1
H 59	12	30	12	10	13	15	16	25	10	13/16	1
H 60	12	30	11	10	11	14	15	22	9	11/14	1
H 61	12	31	12	10	12	12	15	23	10	12/17	1
H 62	12	31	12	10	13	15	15	24	10	11/20	1
H 63	12	32	11	10	13	12	16	23	11	12/15	1
H 64	12	33	11	10	14	12	14	23	10	12/16	1
H 65	13	27	12	10	11	12	16	23	11	15/18	1
H 66	13	27	10	11	13	13	14	22	10	11/19	1
H 67	13	27	11	10	13	12	15	23	10	13/13	1
H 68	13	27	11	10	14	15	15	23	10	14/17	1
H 69	13	28	11	10	15	12	14	23	10	13/20	1
H 70	13	28	12	10	11	15	16	23	10	16/18	1
H 71	13	28	12	10	12	13	15	22	10	12/12	1
H 72	13	28	13	10	10	13	15	24	10	13/13	1
H 73	13	28	13	11	11	12	15	25	11	11/11	1
H 74	13	28	13	13	11	15	15	23	9	11/17	1
H 75	13	28	12	12	12	12	13	25	10	13/18	1
H 76	13	28	11	11	13	12	14	25	10	10/13	1
H 77	13	29	12	11	13	12	16	25	10	12/19	1
H 78	13	29	12	11	14	13	15	25	10	12/13	2
H 79	13	29	12	13	11	12	16	24	9	12/14	1
H 80	13	29	13	10	11	13	14	24	10	12/18	1
H 81	13	29	13	10	13	11	15	24	10	12/14	1
H 82	13	29	13	13	14	12	13	24	10	13/17	1
H 83	13	29	10	10	11	14	15	25	9	13/20	1
H 84	13	29	10	12	14	12	15	23	10	15/19	1
H 85	13	30	11	10	12	12	15	25	10	10/10	1
H 86	13	30	12	10	11	12	15	25	10	11/12	2
H 87	13	30	12	10	14	12	16	23	9	12/14	1
H 88	13	30	12	10	11	14	14	22	10	13/16	1
H 89	13	30	12	10	13	14	14	24	11	11/16	1
H 90	13	30	12	11	11	12	15	23	10	13/13	1
H 91	13	30	12	13	15	13	14	24	10	11/19	1
H 92	13	30	12	10	14	15	16	25	11	12/16	1
H 93	13	30	13	12	11	12	15	24	10	12/12	1
H 94	13	30	13	12	14	12	17	24	10	11/15	1
H 95	13	31	10	11	11	13	16	24	11	14/18	1
H 96	13	31	11	10	13	13	15	24	10	17/20	1
H 97	13	31	11	10	14	14	15	24	10	12/17	1
H 98	13	31	11	12	12	12	13	25	11	14/19	1
H 99	13	31	12	10	13	12	15	23	10	13/20	1
H 100	13	31	12	10	11	12	15	23	10	12/12	1
H 101	13	32	12	11	12	12	15	23	10	12/20	1
H 102	14	27	12	10	13	12	16	23	11	12/16	1
H 103	14	27	12	10	14	12	15	24	10	12/13	2
H 104	14	28	12	10	13	15	14	22	9	11/17	1
H 105	14	28	12	11	12	13	14	26	10	13/18	1
H 106	14	28	12	10	11	14	15	25	11	13/13	1
H 107	14	28	12	11	12	12	15	23	10	12/12	1
H 108	14	28	12	11	11	12	15	24	10	10/10	1
H 109	14	28	13	13	14	12	17	24	10	14/17	1

TABLE 2—Continued.

Haplotype	DYS389I	DYS389II	DYS439	DYS438	DYS392	DYS393	DYS19	DYS390	DYS391	DYS385a/b	No.
H 110	14	29	11	11	13	12	14	24	9	11/22	1
H 111	14	29	11	11	13	11	14	25	10	13/15	1
H 112	14	29	12	10	14	12	15	24	10	14/16	1
H 113	14	29	12	10	14	12	16	24	11	15/17	1
H 114	14	29	12	10	14	12	16	25	11	13/16	1
H 115	14	29	12	10	12	15	15	23	10	17/20	1
H 116	14	29	12	11	12	13	15	24	11	13/13	1
H 117	14	29	12	10	11	13	15	25	12	11/15	1
H 118	14	29	13	10	13	14	17	23	10	12/19	1
H 119	14	30	10	10	13	13	14	23	10	11/21	1
H 120	14	30	11	10	14	14	15	23	10	12/12	1
H 121	14	30	11	11	14	13	15	24	10	13/15	1
H 122	14	30	12	10	13	15	15	23	10	11/20	1
H 123	14	30	12	10	13	12	16	24	10	12/21	1
H 124	14	30	12	12	13	12	14	23	8	13/18	1
H 125	14	30	12	10	11	13	14	23	10	16/19	1
H 126	14	30	12	10	11	13	14	23	10	13/17	1
H 127	14	30	12	13	14	12	13	24	10	13/20	1
H 128	14	30	13	12	14	13	14	22	11	12/18	1
H 129	14	30	13	10	14	14	17	24	11	11/19	1
H 130	14	31	10	10	14	13	14	22	10	11/17	1
H 131	14	31	11	10	14	14	14	22	10	12/17	1
H 132	14	31	12	10	13	12	15	23	10	14/21	1
H 133	14	31	12	10	13	12	14	24	11	12/18	1
H 134	14	32	11	10	14	13	15	25	12	11/15	1
H 135	15	30	12	11	13	12	14	23	10	11/12	1

TABLE 3—Gene diversity and haplotype diversity in different populations.

Locus	This study (<i>n</i> = 141)	Chinese (<i>n</i> = 136)	Japanese (<i>n</i> = 115)	Italian (<i>n</i> = 75)	Caucasian (<i>n</i> = 517)	African (<i>n</i> = 535)	Hispanic (<i>n</i> = 245)	Pakistani (<i>n</i> = 711)	Bangladeshi (<i>n</i> = 72)
DYS389I	0.6508 ⁵	0.6414 ³	0.6611 ⁵	0.5619 ⁴	0.5061 ⁵	0.4931 ⁵	0.5497 ⁵	0.6190 ⁴	0.5967 ⁴
DYS389II	0.7801 ⁸	0.7653 ⁷	0.7940 ⁷	0.7379 ⁶	0.6432 ⁷	0.7373 ⁷	0.6987 ⁶	0.7080 ⁷	0.7633 ⁷
DYS439	0.6152 ⁵	0.6337 ⁴	0.6536 ⁴	0.6650 ⁴	0.6489 ⁵	0.6197 ⁵	0.6462 ⁴	0.7240 ⁶	0.7483 ⁶
DYS438	0.5342 ⁵	0.5136 ⁵	0.5985 ⁶	0.7097 ⁵	0.5486 ⁶	0.5288 ⁵	0.6327 ⁵	0.6490 ⁷	0.6694 ⁵
DYS392	0.7609 ⁶	0.7435 ⁶	0.6884 ⁵	0.5721 ⁴	0.5771 ⁶	0.4126 ⁶	0.6287 ⁶	0.5770 ⁸	0.4661 ⁶
DYS393	0.6429 ⁶	0.6493 ⁵	0.4868 ⁴	0.5892 ⁵	0.3235 ⁶	0.6005 ⁶	0.4481 ⁶	0.6730 ⁶	0.6840 ⁶
DYS19	0.7058 ⁵	0.6619 ⁵	0.6761 ⁵	0.6839 ⁵	0.4248 ⁵	0.7284 ⁵	0.6149 ⁶	0.7100 ⁷	0.6965 ⁵
DYS390	0.7068 ⁶	0.6844 ⁶	0.7571 ⁷	0.7071 ⁶	0.6759 ⁵	0.6015 ⁶	0.6141 ⁶	0.7720 ⁷	0.7852 ⁶
DYS391	0.4869 ⁶	0.4623 ⁵	0.2860 ⁵	0.5084 ³	0.5502 ⁴	0.6013 ⁴	0.5827 ⁴	0.4670 ⁶	0.3447 ³
DYS385a/b	0.8921 ¹⁴	0.8068 ¹⁴	0.9446 ¹³	0.9322 ¹¹	0.7478 ¹⁰	0.8706 ¹⁰	0.8228 ¹¹	—	0.9325 ¹³
HD	0.9998	0.9907	0.9997	0.9996*	0.9946	0.9991	0.9973	0.9900*	0.9996

*A total gene diversity.

The number of superscript in the frequencies: observed number of the allele.
STR, short tandem repeat; HD, haplotype diversity.

References

- John SWM, Weitzner G, Rozen R, Scriver CR. A rapid procedure for extracting genomic DNA from leukocytes. *Nucleic Acids Res* 1991;19:408.
- Schneider S, Roessli D, Excoffier L. A software for population genetics data analysis. Genetic and Biometry Laboratory, Arlequin Ver 2.000. Switzerland: University of Geneva; 2000.
- Nei M. *Molecular evolutionary genetics*. New York: Columbia University Press; 1987.
- Li W, Hu L, Chen S, Qiu N. 10 Y-STRs haplotypes in Chinese. *Forensic Sci Int* 2003;139:85–8.
- Tie J, Wang X, Suzuki Y, Tsukamoto S, Oshida S. Polymorphisms of six Y-chromosome STRs in a Chinese population. *Legal Med* 2004;6:41–6.
- Kido A, Hara M, Kameyama H, Yamamoto Y, Susukida R, Oya M. Y-chromosomal STR loci data in Japanese using the Y-PLEXTM 5 and Y-PLEXTM 6 PCR Amplification Kit. *J Forensic Sci* 2004;49(4):849–51.
- Biondo R, Caglià A, Asili P, D'Agostaro G, D'Agostino G, Spinella A. Typing of 20 Y-chromosome STR in the Italian population. *Forensic Sci Int* 2004;146S:S135–8.
- Sinha SK, Budowle B, Chakrabort R, Paunovic A, Guidry RDV, Larsen C, et al. Utility of the Y-STR typing systems Y-PLEXTM 6 and Y-PLEXTM 5 in forensic casework and 11 Y-STR haplotype database for three major population groups in the United States. *J Forensic Sci* 2004;49(4):691–700.
- Mohyuddin A, Ayub Q, Qamar R, Zerjal T, Helgason A, Mehdi SQ, Tyler-Smith C. Y-chromosomal STR haplotypes in Pakistani populations. *Forensic Sci Int* 2004;118:141–6.
- Dobashi Y, Kido A, Fujitani N, Susukida R, Hara M, Oya M. Y-chromosomal STR haplotypes in a Bangladeshi population. *Legal Med* 2005;7:122–6.

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