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FOR THE RECORD

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Haplotype Frequencies of Four Y-chromosome STR Loci in Chinese Population

POPULATION: Chinese population

KEYWORDS: forensic science, Y chromosome, haplotype, Chinese population, DYS434, DYS435, DYS439, A10, STR population genetics, DNA typing

We investigated the distributions of haplotypes for four Y-STR loci, named as DYS434, DYS438, DYS439, and A10, and developed a multiplex amplified for these Y-STR markers. A total of 100 samples were analyzed. Each sample was amplified utilizing multiplex PCR for four Y-STR loci. The PCR products were analyzed by non-denaturing polyacrylamide gel electrophoresis and visualized by silver staining. A total of 37 different haplotypes were found, and 16 haplotypes were being unique. Gene diversity ranged from 0.3945 at DYS434 to 0.7051 at DYS439. The haplotype diversity, chance of exclusion and discrimination power of four Y-STR loci was 0.9628, SE 0.00429. The results indicated that they were suitable Y linked markers for forensic applications in our population studied.

Y-chromosomal STR loci are of increasing interest in paternity testing, discriminating for sex assault, anthropological and evolutionary studies. We developed a multiplex amplification for four Y chromosomal STR loci, named as DYS434, DYS435, DYS439, and A10, and investigated distributions of haplotypes for four Y-chromosome STR loci in a Chinese population.

EDTA-blood sample were collected from 100 unrelated males of Han population in Chengdu of China. Primer sequences were retrieved on GDB (1). Table 1 listed the sequences of primers of four Y-STR loci.

DNA was extracted using Chelex method (2). Reaction volume of multiplex PCR amplifications was 37.5 μL, containing 2–4 ng human genome DNA, 200 μM each dNTP, 3 μ Taq polymerase, 3.75 μL 10 × buffer, Mg^{2+} 1.5 mM, 1.6 μg/mL BSA, concentration of primers according to the references (3). Amplification reactions were carried out in a Perkin Elmer 9600 (Foster City, CA) with predenaturing for 2 min at 94°C, followed by 38 cycles of denaturing for 50 s at 94°C, annealing for 50 s at 56°C and extension for 25 s at 72°C.

Polyacrylamide gel electrophoresis with continuous buffer system and visualized by staining with silver (4). Human allelic ladders for STR typing were made in-house. Mixing products of PCR with different genotypes constructed the allelic ladders. Alleles were named according to the recommendations of the International Society of Forensic genetics.

TABLE 1—*Sequences of primers of four Y-STR loci.*

Loci	Primer	Sequences of Primer
DYS434	L214-9L	5/CACTCCCTGAGTGCTGGATT
	L214-9R	5/GGAGATGAATGAATGGATGGA
DYS438	L216-15L	5/TGGGGAATAGTTGAACGGTAA
	L216-15R	5/GTGGCAGACGCCTATAATCC
DYS439	L214-18L	5/TCCTGAATGGTACTTCCTAGGTTT
	L214-18R	5/GCCTGGCTTGGAATTCTTTT
A10	A	5/ATAAATGGAGATAGTGGGTGGATT
	B	5/CCTGCCATCTCTATTTATCTTGCATATA

TABLE 2—*Allele frequencies and gene diversities at four Y-chromosome STR loci Chinese population.*

Allele	DYS434	DYS438	DYS439	A10
9	0.0988	0.0123		
10	0.7654	0.7531	0.1235	
11	0.0494	0.2099	0.1074	0.0288
12	0.0864	0.0247	0.2963	0.3462
13			0.1605	0.3654
14			0.0123	0.2404
15				0.0192
Gene Diversity	0.3945	0.3880	0.7051	0.6876
SE	0.0454	0.0391	0.0177	0.0113

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TABLE 3—*The haplotypes of four Y-chromosome STR loci in Chinese population.*

Haplotype	DYS434	A10	DYS438	DYS439	Frequency, %
H1	9	12	10	10	3
H2	9	12	10	11	2
H3	9	12	11	10	1
H4	9	12	11	11	1
H5	9	12	11	12	1
H6	10	11	10	11	1
H7	10	11	12	11	2
H8	10	12	9	11	1
H9	10	12	10	10	1
H10	10	12	10	11	5
H11	10	12	10	12	6
H12	10	12	10	13	2
H13	10	12	11	11	1
H14	10	12	11	12	1
H15	10	12	11	13	2
H16	10	13	10	10	2
H17	10	13	10	11	8
H18	10	13	10	12	11
H19	10	13	10	13	4
H20	10	13	11	11	1
H21	10	13	11	12	6
H22	10	13	11	13	4
H23	10	14	9	12	1
H24	10	14	10	10	2
H25	10	14	10	11	5
H26	10	14	10	12	6
H27	10	14	10	13	4
H28	10	14	11	11	2
H29	10	14	11	13	1
H30	10	15	10	12	1
H31	10	15	11	13	1
H32	11	12	10	10	1
H33	11	12	10	11	2
H34	11	14	10	11	1
H35	12	12	10	11	4
H36	12	12	10	12	2
H37	12	14	10	11	1
Diversity			0.9628		
SE			0.00429		

The gene diversity and standard error were calculated in accordance with Hou's method (5). The formula calculating the gene diversity: $h = n(l - \sum X^2)/(n - l)$, where n is the number of population, h is the gene diversity, x is the frequencies of allele. The formula calculating Standard error (S.E.): $S.E. = \{2\{\sum X^3 - (\sum X^2)^2/n\}^{1/2}$.

The multiplex amplification for four Y-STR loci was precise and reproducible genotype for four Y-STR loci. Development of Y-chromosome specific polymorphisms will be of great benefit in analyzing mixed DNA samples, in investigating sexual assaults, and kinship testing of paternal relative of forensic science (6). Our results suggested that gene diversity of DYS434, DYS438, A10 and DYS439 was 0.3945, 0.3880, 0.6876 and 0.7051, respectively. DYS439 showed the highest diversity value. Allele frequencies for four Y-STR loci and gene diversity values are shown in Table 2. Haplotype diversity of four loci was 0.9628, and SE was 0.00429. This means that chance of exclusion and discrimination power are 0.9628. The result showed that they were suitable Y linked markers for forensic applications in our studied population. Table 2 haplotypes constructed from three Y-STR loci and the haplotype diversity was shown in Table 3.

The complete data can be obtained from the authors on request to: jiqiangl821@163.com.

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