

Announcement of Population Data
Allele frequencies and haplotypes of 12 Y-STR loci for the
local Chinese population in Hong Kong

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Abstract

Haplotype frequencies were established for 12 Y-chromosome STR loci, including all loci recommended by Scientific Working Group on DNA Analysis Methods Y-STR Subcommittee (DYS391, *DYS389I/II*, *DYS439*, *DYS393*, *DYS390*, *DYS385a/b*, *DYS438*, *DYS19* and *DYS392*) plus *DYS437*, in the local Chinese population in Hong Kong. In a sample of 481 unrelated males, it was possible to define 424 different haplotypes of which 388 were unique, 26 was found in two individuals, 2 were shared in three individuals, 5 were shared in four individuals and 3 were shared in five individuals. The allele diversity values for each locus ranged from 0.4273 (*DYS438*) to 0.9555 (*DYS385a/b*). The observed haplotype diversity value and discrimination capacity were 0.9992 and 0.8815, respectively. In a genetic study of these unrelated males, triple alleles were found at the *DYS358* locus in six individuals. The combined Y-chromosome STR polymorphisms provide a powerful discrimination tool for routine forensic applications.

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Keywords: Y-chromosome; Short tandem repeats (STRs); Haplotypes; Chinese population

Population: 447 buccal swabs and 34 blood samples of total 481 unrelated individuals of the local Chinese in Hong Kong.

DNA extraction: DNA were extracted by Chelex extraction method [1] and QIAamp DNA Blood Mini Kit (Qiagen). The concentrations of the DNA were quantified by QuantiBlot[®] Human DNA Quantitation Kit (Perkin-Elmer), Quantifiler[™] Y Human Male DNA Quantification Kit (Applied Biosystems) or PicoGreen[®] dsDNA quantitation reagent (Molecular Probes).

PCR: 1.5 ng target DNA was amplified using the commercial kit PowerPlex[®] Y System (Promega), following manufacturer's instruction.

Cycling conditions: 95 °C for 11 min and 96 °C for 1 min; followed by 10 cycles of 94 °C for 30 s, 60 °C for 30 s and 70 °C for 45 s, 20 cycles of 90 °C for 30 s, 58 °C for 30 s and 70 °C for 45 s, and a final extension of 60 °C for 30 min and soak in 4 °C in a Perkin-Elmer GeneAmp[®] PCR System 9700 Thermal Cycler.

Typing: ABI Prism[™] 310 or 3100 Genetic Analyzer (Applied Biosystems) using the recommended protocol

(PowerPlex[®] Y System, Promega), electrophoresis results were analyzed by GeneScan[®] and Genotyper[®] software (Applied Biosystems) with the PowerTyper[™] Y Macro (Promega Corporation).

Results: See Tables 1–4.

Quality control: Quality assurance standards as stipulated by SWGDAM were followed.

Analysis of data: Haplotype and allele frequencies were estimated by gene counting. Observed gene and haplotype diversities were calculated according to Nei [2] with the formula: $h = n(1 - \sum x^2)/(n - 1)$, where h is the allele diversity or haplotype diversity, n represents the number of individuals, and x is the allele or the haplotype frequencies in a given population sample. The discrimination capacity was the percentage proportion of different haplotypes.

Other remarks: Tables 1 and 2 show the allele frequencies distribution of each locus. A total of 424 different haplotypes were identified of which 388 were observed only once (Table 4). The observed haplotype diversity was 0.9992, and the discrimination capacity was 0.8815.

Allele diversities (Table 3) were calculated from the allele frequencies. In this study, *DYS385a/b* and *DYS389II* were the most informative loci with overall allele diversity of 0.9555 and

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Table 1
Allele frequencies at 10 Y-STR loci in a local Chinese population in Hong Kong^a

Allele	DYS391	DYS389I	DYS439	DYS389II	DYS438	DYS437	DYS19	DYS392	DYS393	DYS390
6	0.0042									
7								0.0021		
8	0.0021									
9	0.0208				0.0083					
10	0.6819		0.0333		0.7089			0.0021	0.0021	
11	0.2827	0.0146	0.2827		0.2661			0.0728	0.0021	
12	0.0083	0.5405	0.4886		0.0125			0.0561	0.4449	
13		0.2911	0.1726		0.0042	0.0042	0.0312	0.4116	0.3077	
14		0.1497	0.0229			0.6112	0.2308	0.4324	0.2162	
15		0.0042				0.3763	0.4657	0.0229	0.0249	
16						0.0083	0.2079		0.0021	
17							0.0624			
18							0.0021			
19										
20										
21										0.0021
22										0.0561
23										0.3846
24										0.3347
25										0.2079
26				0.0166						0.0146
27				0.0520						
28				0.3555						
29				0.2807						
30				0.1996						
31				0.0894						
32				0.0062						

^a $n = 481$.

0.7455, respectively. DYS391, DYS438 and DYS437 were the least informative loci with allele diversity below 0.5.

Most of the commonly used Y-STR markers are single-copy loci, with the notable exception of the polymorphic locus

Table 2
Allele frequencies at DYS385 in a local Chinese population in Hong Kong^a

Allele	DYS385	Allele	DYS385	Allele	DYS385
9–18	0.0021	12–18	0.0312	14–14	0.0042
9–19	0.0021	12–19	0.0395	14–15	0.0042
10–11	0.0021	12–20	0.0353	14–16	0.0042
10–16	0.0021	12–21	0.0042	14–17	0.0062
10–17	0.0021	12–22	0.0021	14–18	0.0333
10–18	0.0021	12–23	0.0083	14–19	0.0125
11–11	0.0104	12–13–18	0.0021	14–20	0.0042
11–12	0.0416	12–13–20	0.0021	14–21	0.0062
11–13	0.0062	13–13	0.1270	14–22	0.0021
11–15	0.0021	13–14	0.0416	15–15	0.0042
11–16	0.0042	13–15	0.0021	15–16	0.0083
11–17	0.0125	13–16	0.0187	15–17	0.0021
11–18	0.0062	13–17	0.0603	15–18	0.0125
11–19	0.0125	13–18	0.0769	15–19	0.0187
11–20	0.0021	13–19	0.0707	15–20	0.0083
11–21	0.0062	13–20	0.0395	15–21	0.0062
12–12	0.0062	13–21	0.0229	16–17	0.0021
12–13	0.0395	13–22	0.0125	16–18	0.0021
12–14	0.0083	13–23	0.0042	16–19	0.0083
12–15	0.0125	13–16–17	0.0021	16–20	0.0062
12–16	0.0229	13–17–18	0.0042	17–17	0.0021
12–17	0.0249	13–18–19	0.0021	20–20	0.0021

^a $n = 481$.

DYS385. Two alleles are often observed in DYS385 locus since two fragments are commonly found in Y-chromosome. In this study, we found 6 out of the 481 samples (1.25%) tested contained triple alleles in DYS385, representing the presence of duplicated regions of the Y-chromosome [3].

Of the 3 haplotypes shared by five individuals (Table 4), the haplotype no. 318 was found to have three matches (frequency = 0.92) in an Asian population sample of 325 haplotypes (Promega), and eight matches (frequency = 0.46) in an Asian population sample of 1729 haplotypes from Y-chromosome haplotype reference database (YHRD, <http://www.ystr.org>). Seven matches (frequency = 0.40) were also found for the haplotype no. 59 compared to the YHRD database. It should be noted that there was no match for haplotype no.

Table 3
Observed alleles and allele diversities for the analyzed loci

Y-STR	No. of observed alleles	Allele diversity
DYS391	6	0.4555
DYS389I	5	0.6017
DYS439	5	0.6513
DYS389II	7	0.7455
DYS438	5	0.4273
DYS437	4	0.4858
DYS19	6	0.6832
DYS392	7	0.6376
DYS393	7	0.6614
DYS390	6	0.6949
DYS385	66	0.9555

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