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Haplotype distribution of Y-STR loci in Beijing population

Y.C. Liu*, J. Yang, J. Wang, H. Tang, Z.Y. Huo

Forensic Medical Examination and Identification Centre of Beijing Public Security Bureau, 1 Longgang Road, Qinghe District, Beijing 100085, China

Abstract. The use of Y chromosome markers is of special interest because of their male-specificity. Y-STR plays a very important role in forensic science, especially in sexual assault cases when another person masks the DNA of one male and deficiency paternity cases involving male offspring. In our study, we investigated the haplotype distribution of 10 Y-STR loci (DYS393, DYS19, DYS389 II, DYS389 I, DYS390, DYS391, DYS385, DYS439, DYS438, DYS392) in Beijing population.

Keywords: Y-STR; Haplotype; Beijing population

1. Materials and methods

Blood specimens were collected from 123 unrelated males, 100 father–son pairs, 50 male sibling pairs. DNA was extracted using 5% Chelex 100. PCR amplification was performed using Y-Plex™ 6 and Y-Plex™ 5 kit in a 25-μl reaction volume, including 1–5 ng genomic DNA, 5.0 μl primer, 0.5 μl (5 u/μl) AmpliTag Gold™. PCR cycling conditions referred to the kit manual. PCR products ran on ABI 377 DNA sequencer.

2. Results and discussions

Allele frequency distribution, Dp and GD of the 10 Y-STR loci of Beijing population were obtained (see Table 1). Table 2 showed that the more the loci that we used the higher Dp and GD value that we got.

The results of the father–son pairs and sibling pairs proved that the 10 Y-STR loci we studied followed male genetic rules. There were two variances that happened in DYS389 II (29 → 28) and DYS385 (15/17 → 15/18) loci when the father transmitted Y chromosome to the son. So we suggested that more than three different Y-STR typing can lead to exclusion.

* Corresponding author. Tel.: +86-10-62909016; fax: +86-10-62902582.
E-mail address: fyzhx6688@163.com (Y.C. Liu).
URL: www.Fmeicbjpsb.sohu.com.

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Table 1
The frequency of 10 Y-STR loci in unrelated males from Beijing ($n = 123$)

Allele	DYS393	DYS19	DYS389II	DYS390	DYS391	DYS389I	DYS439	DYS438	DYS392	DYS385			
6					0.0053								
7									0.0081	9–18	0.0053	12–19	0.0481
8										10–13	0.0053	12–20	0.0214
9					0.0642		0.0081	0.0163		10–15	0.0053	12–22	0.0107
10	0.0053				0.7701		0.0488	0.6667	0.0081	10–16	0.0107	13–13	0.0481
11	0.0053				0.1444	0.0081	0.3821	0.2520	0.1220	10–17	0.0053	13–14	0.0428
12	0.5294	0.0053			0.0160	0.5203	0.4228	0.0488	0.1060	10–18	0.0214	13–15	0.0160
13	0.2674	0.0374				0.2764	0.1301	0.0163	0.3496	10–19	0.0053	13–16	0.0214
14	0.1551	0.3102				0.1707			0.3659	10–20	0.0053	13–17	0.0267
15	0.0267	0.3743				0.0244	0.0081		0.0325	11–11	0.0214	13–18	0.0642
16	0.0107	0.2193							0.0081	11–12	0.0535	13–19	0.0428
17		0.0481								11–13	0.0214	13–20	0.0374
18		0.0053								11–14	0.0053	14–14	0.0053
19										11–15	0.0053	14–16	0.0053
20										11–16	0.0160	14–17	0.0107
21				0.0160						11–17	0.0214	14–18	0.0267
22				0.0856						11–18	0.0160	14–19	0.0107
23				0.3476						11–19	0.0214	14–21	0.0053
24				0.3743						11–20	0.0053	15–17	0.0053
25				0.1551						11–21	0.0053	15–19	0.0160
26			0.0107	0.0214						12–12	0.0374	15–20	0.0107
27			0.0642							12–13	0.0214	15–21	0.0160
28			0.3262							12–14	0.0428	15–22	0.0053
29			0.3262							12–15	0.0053	16–16	0.0053
30			0.1872							12–16	0.0374	16–17	0.0053
31			0.0802							12–17	0.0428	17–20	0.0053
32										12–18	0.0428		
33			0.0053										
DP	0.6233	0.7118	0.7414	0.7070	0.3817	0.6231	0.6558	0.4891	0.8371		0.9676		
GD	0.6267	0.7156	0.7456	0.7108	0.3838	0.6282	0.6612	0.4931	0.8440		0.972		

Table 2
The relationship between the number of loci and DP, GD

Y-STR loci	Haplotype no.	DP	GD
DYS393 + DYS19	17	0.8792	0.8864
DYS393 + DYS19 + DYS389II	45	0.9527	0.9605
DYS393 + DYS19 + DYS389II + DYS390	76	0.9787	0.9867
DYS393 + DYS19 + DYS389II + DYS390 + DYS391	91	0.9837	0.9917
DYS393 + DYS19 + DYS389II + DYS390 + DYS391 + DYS385	118	0.9912	0.9993
DYS393 + DYS19 + DYS389II + DYS390 + DYS391 + DYS385 + DYS389I	119	0.9913	0.9995
DYS393 + DYS19 + DYS389II + DYS390 + DYS391 + DYS385 + DYS389I + DYS439 + DYS438 + DYS392	121	0.9916	0.9997

Two male DNA were diluted to 0.5 ng/μl, 0.1 ng/μl, 50 pg/μl, 20 pg/μl, 10 pg/μl, 5 pg/μl, 1 pg/μl. Successful Y-STR typing results were obtained when DNA was more than 5 pg.

Female and male DNA were mixed as the following ratio (female/male): 6000:1, 4000:1, 3000:1, 2000:1, 1000:1, 500:1, 100:1, 50:1. Y-STR typing of male was obtained when the ratio was less than 2000:1.

Knowledge about the geographical frequency distribution of Y-STR haplotypes is important for the forensic community. The information of the 10 Y-STR loci in Beijing population provides additional data to autosomal STRs.