TECHNICAL NOTE

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Japanese Population DNA Typing Data for the Loci LDLR, GYPA, HBGG, D7S8, and GC

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ABSTRACT: Population studies on the five loci LDLR, GYPA, HBGG, D7S8, and GC (PM loci) were carried out in a sample of 366 unrelated Japanese individuals living in Gifu Prefecture (central region of Japan) using the AmpliType® PM PCR Amplification and Typing kit (Perkin Elmer). For all loci, no significant deviations from Hardy-Weinberg equilibrium could be found in our Japanese population sample. The combined mean exclusion chance and power of discrimination for the PM loci were 0.68 and 0.993, respectively. The Japanese and Chinese population data were similar for the all loci. The Japanese and Korean population data were similar for all loci other than D7S8. Significant differences were observed between the Japanese population data and the 16 other population data compared for 3 loci or more.

KEYWORDS: forensic science, DNA typing, polymerase chain reaction, LDLR, GYPA, HBGG, D7S8, GC, Japan, population data

The loci LDLR (low density lipoprotein receptor) (1), GYPA (glycophorin A) (2), HBGG (hemoglobin G gammaglobin) (3), D7S8 (4), and GC (group specific component) (5) are amplified with the AmpliType PM PCR Amplification and Typing kit. The five loci(PM loci) listed are typed simultaneously in a single reverse dot blot strip containing immobilized allele specific probes. This paper presents the population data for the PM loci in a Japanese population sample and an evaluation of the potential forensic utility of the PM loci in Japanese. In addition, the Japanese population data are compared with data from 18 other populations (6–16).

Materials and Methods

Sample Preparation

Blood samples were obtained from 366 unrelated Japanese individuals living in Gifu Prefecture (central region of Japan). DNA was extracted using the phenol-chloroform method (17).

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Typing

The loci were amplified and typed using the AmpliTypePM PCR Amplification and Typing kit (Perkin Elmer) according to the manufacture's protocol.

Statistical Analysis

To estimate if the Japanese population sample examined in this study conforms to the Hardy-Weinberg equilibrium, the conventional chi-square test between observed and expected genotype frequencies was carried out for each locus. The mean exclusion chance (MEC) was calculated using the computer programe described by Ohno et al. (18). The power of discrimination (PD), expected heterozygosity (H-exp) and the standard error (SE) were calculated according to Fisher (19) and Edwards et al. (20), respectively. Examinations for population sample homogeneity were done by the chi-square tests of 2 × C contingency tables.

Results and Discussion

The distribution of genotypes and allele frequencies for the PM loci in the Japanese population sample are shown in Table 1. For all loci, no significant deviations from Hardy-Weinberg equilibrium could be found.

Table 2 summarizes various statistical parameters of forensic interest calculated for the PM loci in our Japanese population sample. The PM loci ranged in expected heterozygosity from 0.30 (LDLR) to 0.62 (GC), mean exclusion chance from 0.13 (LDLR) to 0.34 (GC), and power of discrimination from 0.47 (LDLR) to 0.78 (GC). The combined mean exclusion chance and power of discrimination for the PM loci were 0.68 and 0.993, respectively. The present data demonstrate that the PM typing is a useful technique for paternity testing and individual identification in the Japanese population.

A quantitative comparison of allele frequencies for the PM loci between this study and 18 other population studies (6–16) is shown in Table 3. No significant differences were observed between Japanese and Chinese population data for all loci. Except for D7S8, no significant differences were observed between Japanese and Korean population data. The difference for D7S8 between Japanese and Korean population data may be due to genetic differences or to sample sizes as described by Woo and Budowle (7) for D1S80 locus. Significant differences were observed between our Japanese

TABLE 1—*Distribution of genotypes and allele frequencies for PM loci in 366 unrelated Japanese.*

Locus	Genotype	No. of obs. (%)	No. of exp. (%)	Allele frequencies	
LDLR	AA	13 (3.6)	12.8 (3.5)	LDLR* A=	0.187
	AB	111 (30.3)	111.4 (30.4)	LDLR* B=	0.813
	BB	242 (66.1)	241.8 (66.1)		
$\chi^2 = 0.004$, $df = 1$, $0.8 < P < 0.9$					
Locus	Genotype	No. of obs. (%)	No. of exp. (%)	Allele frequencies	
GYPA	AA	111 (30.3)	117.1 (32.0)	GYPA* A=	0.566
	AB	192 (52.5)	179.9 (49.2)	GYPA* B=	0.434
	BB	63 (17.2)	69.1 (18.9)		
$\chi^2 = 1.67$, $df = 1$, $0.1 < P < 0.2$					
Locus	Genotype	No. of obs. (%)	No. of exp. (%)	Allele frequencies	
HBGG	AA	34 (9.3)	30.7 (8.4)	HBGG* A=	0.290
	AB	144 (39.3)	150.6 (41.1)	HBGG* B=	0.710
	AC	0 (0.0)	0.0 (0.0)	HBGG* C=	0.000
	BB	188 (51.4)	184.7 (50.5)		
	BC	0 (0.0)	0.0 (0.0)		
	CC	0 (0.0)	0.0 (0.0)		
$\chi^2 = 0.70$, $df = 1$, $0.3 < P < 0.5$					
Locus	Genotype	No. of obs. (%)	No. of exp. (%)	Allele frequencies	
D7S8	AA	139 (38.0)	140.2 (38.3)	D7S8* A=	0.619
	AB	175 (47.8)	172.7 (47.2)	D7S8* B=	0.381
	BB	52 (14.2)	53.2 (14.5)		
$\chi^2 = 0.07$, $df = 1$, $0.7 < P < 0.8$					
Locus	Genotype	No. of obs. (%)	No. of exp. (%)	Allele frequencies	
GC	AA	25 (6.8)	25.2 (6.9)	GC* A=	0.262
	AB	94 (25.7)	98.9 (27.0)	GC* B=	0.515
	AC	48 (13.1)	42.8 (11.7)	GC* C=	0.223
	BB	105 (28.7)	97.1 (26.5)		
	BC	73 (19.9)	83.9 (22.9)		
	CC	21 (5.7)	18.1 (4.9)		
$\chi^2 = 3.41$, $df = 3$, $0.3 < P < 0.5$					

TABLE 2—*Forensic efficiency values of PM loci in the Japanese population sample.*

Locus	H-obs	H-exp ± SE	MEC	PD
LDLR	0.30	0.30 ± 0.024	0.13	0.47
GYPA	0.52	0.49 ± 0.026	0.19	0.62
HBGG	0.39	0.41 ± 0.026	0.16	0.57
D7S8	0.48	0.47 ± 0.026	0.18	0.61
GC	0.59	0.62 ± 0.025	0.34	0.78
Combined			0.68	0.993

H-obs: observed heterozygosity, H-exp: expected heterozygosity, SE: standard error, MEC: mean exclution chance, PD: power of discrimination.

TABLE 3—*P-values for pairwise comparisons of our Japanese allele frequencies* (n = *366) with 18 other population data.*

Population	*n*	LDLR	GYPA	HBGG	D7S8	GC	References
Chinese	105	$0.1 < P < 0.2$	$0.3 < P < 0.5$	$0.3 < P < 0.5$	$0.3 < P < 0.5$	$0.1 < P < 0.2$	(6)
Korean	116	$0.3 < P < 0.5$	$0.3 < P < 0.5$	$0.99 < P$	$0.001 < P < 0.01$	$0.5 < P < 0.7$	(7)
U.S. Caucasian	148	$P < 0.001$	$0.5 < P < 0.7$	$P < 0.001$	$0.7 < P < 0.8$	$P < 0.001$	(8)
Swiss Caucasian	100	$P < 0.001$	$0.3 < P < 0.5$	$P < 0.001$	$0.3 < P < 0.5$	$P < 0.001$	(9)
Dutch Caucasian	155	$P < 0.001$	$0.1 < P < 0.2$	$P < 0.001$	$0.95 < P < 0.99$	$P < 0.001$	(10)
Southeastern Hispanic	94	$P < 0.001$	$0.3 < P < 0.5$	$P < 0.001$	$0.3 < P < 0.5$	$P < 0.001$	(8)
Southwestern Hispanic	96	$P < 0.001$	$0.02 < P < 0.05$	$P < 0.001$	$0.1 < P < 0.2$	$P < 0.001$	(8)
African American	145	$0.1 < P < 0.2$	$0.01 < P < 0.02$	$P < 0.001$	$0.8 < P < 0.9$	$P < 0.001$	(8)
Arab	94	$P < 0.001$	$0.2 < P < 0.3$	$P < 0.001$	$0.3 < P < 0.5$	$P < 0.001$	(11)
Galicia	143	$P < 0.001$	$0.01 < P < 0.02$	$P < 0.001$	$0.05 < P < 0.1$	$P < 0.001$	(12)
Spain	132	$P < 0.001$	$0.1 < P < 0.2$	$P < 0.001$	$0.2 < P < 0.3$	$P < 0.001$	(12)
Coimbra	119	$P < 0.001$	$0.8 < P < 0.9$	$P < 0.001$	$0.1 < P < 0.2$	$P < 0.001$	(12)
Hungarian	163	$P < 0.001$	$0.3 < P < 0.5$	$P < 0.001$	$0.5 < P < 0.7$	$P < 0.001$	(13)
Italian	100	$P < 0.001$	$0.1 < P < 0.2$	$P < 0.001$	$0.1 < P < 0.2$	$P < 0.001$	(14)
North Bavarian	150	$P < 0.001$	$0.5 < P < 0.7$	$P < 0.001$	$0.5 < P < 0.7$	$P < 0.001$	(15)
Navajo	81	$P < 0.001$	$P < 0.001$	$0.8 < P < 0.9$	$0.5 < P < 0.7$	$P < 0.001$	(16)
Pueblo	103	$P < 0.001$	$P < 0.001$	$0.3 < P < 0.5$	$0.001 < P < 0.01$	$P < 0.001$	(16)
Sioux	64	$P < 0.001$	$P < 0.001$	$0.01 < P < 0.02$	$P < 0.001$	$P < 0.001$	(16)

population data and 16 other population data for 3 loci or more. The differences for the PM loci between Japanese and 16 other population data may be due to genetic differences. The PM loci in 18 population data ranged in combined mean exclusion chance (MEC) from 0.65 (Navajo) to 0.72 (Southeastern Hispanic) and combined power of discrimination (PD) from 0.991 (Navajo) to 0.996 (Southeastern Hispanic). These data demonstrate that PM typing is a useful technique for forensic identity in these populations.

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