Allele Frequency Distribution of Five Loci (LDLR, GYPA, HBGG, D7S8 and GC) in a Japanese Population

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Introduction

Recently, a kit for the simultaneous genotyping of low density lipoprotein receptor (LDLR)[1], glycophorin A (GYPA)[2], hemoglobin G gammaglobin (HBGG)[3], D7S8 [4] and group specific component (GC)[5] has been commercially available which is based on the techniques of PCR amplification and reverse dot blot hybridization. With this new kit we have determined the distribution of genotypes and allele frequencies of the above five loci in a Japanese population. Moreover, we have investigated the relationship between MN blood types and GYPA genotypes and the relationship between serum GC subtypes and GC genotypes.

Materials and Methods

Blood samples were collected from 257 unrelated Japanese individuals.

DNA was extracted using the SMI TEST kit (Sumitomo Kinzoku). Genotyping of the LDLR, GYPA, HBGG, D7S8 and GC loci was performed using the AmpliType PM PCR Amplification and Typing kit (Perkin Elmer) according to the protocol recommended by the manufacturer.

MN blood types were determined by agglutination with the use of anti-M and -N sera (Ortho).

Serum GC was subtyped by polyacrylamide gel isoelectric focusing followed by immunoblotting [6].

Results and Discussion

The distributions of genotypes and allele frequencies of the five loci LDLR, GYPA, HBGG, D7S8 and GC are shown in Tables 1 and 2. The population data at each locus fitted

the Hardy-Weinberg law. The cumulative probability of paternity exclusion for the 5 loci was calculated at 0.690. The cumulative discrimination power for the 5 loci was as high as 0.992.

Table 1. Distribution of genotypes of LDLR, GYPA, HBGG, D7S8 and GC in 257 unrelated Japanese

Genotype	LDLR	GYPA	HBGG	D7S8	GC
AA	0.027	0.335	0.144	0.377	0.066
AB	0.237	0.494	0.455	0.486	0.237
BB	0.735	0.171	0.401	0.136	0.257
AC			0.000		0.121
BC			0.000		0.253
CC			0.000		0.066

Table 2. Distribution of allele frequencies of LDLR, GYPA, HBGG, D7S8 and GC in 257 unrelated Japanese

Allele	LDLR	GYPA	HBGG	D7S8	GC
A	0.146	0.582	0.372	0.621	0.245
B	0.854	0.418	0.628	0.379	0.502
C			0.000		0.253

Table 3 shows the relationship between MN blood types and GYPA genotypes in 257 Japanese. A complete correspondence of M = GYPA A and N = GYPA B was confirmed.

Table 3. Relationship between MN blood types and GYPA genotypes in 257 unrelated Japanese

MN blood type	GYPA genotype		
	AA	AB	BB
M	86	0	0
MN	0	127	0
N	0	0	44
Total	86	127	44

Table 4 shows the relationship between serum GC subtypes and GC genotypes in 200 Japanese. 187 out of 200 individuals were common subtypes and a correspondence of GC*2 = GC A, GC*1F = GC B and GC*1S = GC C was observed. However, the remaining 13 individuals were variant types; GC 2-1A2 and GC 2-1A9 were GC AB, GC 1F-1A2 and GC 1A3-1A9 were GC

BB, GC 1S-1A2 was GC BC. Thus, GC*1A2, GC*1A3 and GC*1A9 corresponded to GC B as GC*1F did.

Table 4. Relationship between serum GC subtypes and GC genotypes in 200 unrelated Japanese

Serum GC subtype	GC genotype					
	AA	AB	AC	BB	BC	CC
2	13	0	0	0	0	0
2-1F	0	43	0	0	0	0
2-1S	0	0	27	0	0	0
1F	0	0	0	46	0	0
1F1S	0	0	0	0	47	0
1S	0	0	0	0	0	11
2-1A2	0	3	0	0	0	0
2-1A9	0	1	0	0	0	0
1F-1A2	0	0	0	4	0	0
1A3-1A9	0	0	0	1	0	0
1S-1A2	0	0	0	0	4	0
Total	13	47	27	51	51	11

The AmpliType PM PCR kit system permits combined genotyping of LDLR, GYPA, HBGG, D7S8 and GC on a single dot blot strip. The technique is simple, rapid and therefore recommendable to be routinely used for individual identification as well as paternity testing. Because of the frequent occurrence of various GC variants, however, the GC genotyping by the present kit cannot be substituted for the serum GC subtyping.

References

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