

DISTRIBUTION OF HLA - DQ α ALLELES IN DEOXYRIBONUCLEIC ACID (DNA)
FROM CAUCASIAN AND BLACK POPULATIONS OF MARION COUNTY, INDIANA, USA

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The frequency in the population of the genotype markers, found in the evidentiary sample is important in associating an individual as a donor to the crime scene. Among the genotype markers used in forensic science, the HLA-DQ α and the highly polymorphic VNTR (variable number of tandem repeats) loci have the most discrimination power for eliminating a suspect (Budowle, et. al., 1990). However, in those cases where the suspect cannot be excluded as a likely contributor to the crime scene, it is essential to determine the probability of other individuals in the population which can be the potential donor of the biological evidentiary sample. For that matter, the data on the frequency of alleles at HLA-DQ α and VNTR loci are required for statistical analysis. We report in this paper the results on the distribution of alleles at HLA-DQ α locus in DNA from black and caucasian population of Marion County, Indianapolis, Indiana. The data on VNTR loci of these populations will be presented elsewhere.

The blood samples were collected from 203 black and 186 caucasian, all residing in Marion County, Indianapolis, Indiana. About 700 μ L liquid blood was frozen at -80 $^{\circ}$ C for extracting DNA according to the procedure described in FBI Manual (1992). Quantitation of DNA was done spectrophotometrically by recording the absorbance at 260, 270, 280 and 300 nm.

HLA-DQ α genotyping: Two μ L of extracted DNA was used to amplify the specific segment of DNA by polymerase chain reaction (PCR) following the procedure recommended by Cetus (1991). The amplification was performed using Taq polymerase enzyme in a programmed Perkin-Elmer thermal cycler, with cycle setting as follows: denaturation at 94 $^{\circ}$ C for 60 seconds, annealing at 60 $^{\circ}$ C for 30 seconds and extension at 72 $^{\circ}$ C for 30 seconds; the total cycles being 32. With each batch negative and positive controls were included to determine the authenticity of the final product. The amplified DNA was used for HLA-DQ α genotyping as narrated in Cetus Amplitype kit (1991). Tables 1 and 2 depict the observed HLA-DQ α alleles frequencies respectively of black and caucasian population along with the expected frequencies computed by Chi-square method based on the assumption of Hardy-Weinberg equilibrium (Mange and Mange, 1990). It is obvious from Figure 1 that HLA-DQ α alleles distribution in black and caucasian population markedly differed from each other. However, despite these differences, both populations seem to be in conformity with the Hardy-Weinberg Equilibrium. The large probability of Chi-square values suggest that the deviation between the observed and expected genotype frequencies (Table 1,2) can be assigned to chance variance alone. The low proportion of homozygotes in both populations indicates that there were no significant non-random mating sub-populations within these ethnic populations and that there were no undetected alleles in these populations (Cetus, 1991).

Our data on the frequencies of HLA-DQ α alleles distribution in black and caucasian are quite similar to those reported by FBI (1992) and Cetus Corporation (1991) for ethnic populations. From these data it is thus possible to compute the proportion of each genotype which could be expected in a known population.

1. Budowle, B., Baechtel, F.S., Giusti, A.M. and Monson, K.L., "Applying Highly Polymorphic Variable Number of Tandem Repeats Loci Genetic Markers to Identity Testing", *Clinical Biochemistry*, Vol. 23, pp. 287-293 (1990).
2. Budowle, B., Monson, K.L., Anoe, K.S., Baechtel, F.S., Bergman, D.L., Buel, E., Campbell, P.A., Clement, M.E., Corey, H.W., Davis, L.A., Dixon, A., Fish, P., Giusti, A.M., Grant, T.L., Gronert, T.M., Hoover, D.M., Jankowski, L., Kilgore, A.J., Kimoto, W., Landrum, W.H., Leone, H., Longwell, C.R., MacLaren, D.C., Medlin, L.E., Nerveson, S.D., Pierson, M.L., Pollock, J.M., Raquel, R.J., Reznicek, J.M., Rogers, G.S., Smerick, J.E. and Thompson, R.M., "A Preliminary Report on Binned General Population Data on Six VNTR Loci in Caucasians, Blacks, and Hispanics from United States", *Crime Laboratory Digest*, Vol. 18 No. 1, pp. 9-26 (1991).
3. Cetus Amplitype User Guide, Cetus Corporation, Emeryville, California, 1991.

Table 1 HLA DQ Alpha Genotype Distribution in Caucasian Population of Marion County, Indiana

HLA DQ Alpha GENOTYPE	OBSERVED (N = 185) ^a		EXPECTED (N = 185.3) ^a	
1.1, 1.1	4 ^b	(.022) ^c	2.8	(.015) ^c
1.1, 1.2	7	(.038)	10.9	(.059)
1.1, 1.3	2	(.011)	3.1	(.017)
1.1, 2	4	(.022)	5.8	(.031)
1.1, 3	11	(.059)	8.1	(.044)
1.1, 4	14	(.076)	12.3	(.066)
1.2, 1.2	10	(.054)	10.5	(.057)
1.2, 1.3	8	(.043)	6.0	(.032)
1.2, 2	11	(.059)	11.2	(.060)
1.2, 3	15	(.081)	15.5	(.083)
1.2, 4	27	(.146)	23.6	(.127)
1.3, 1.3	0	0	0.8	(.004)
1.3, 2	3	(.016)	3.2	(.017)
1.3, 3	1	(.005)	4.4	(.024)
1.3, 4	11	(.059)	6.7	(.036)
2, 2	1	(.005)	3.0	(.016)
2, 3	13	(.071)	8.3	(.045)
2, 4	14	(.076)	12.6	(.068)
3, 3	5	(.027)	5.7	(.031)
3, 4	15	(.081)	17.5	(.094)
4, 4	9	(.049)	13.3	(.072)

a: N = Number of people typed.

b: number of each genotype.

c: frequency of each genotype.

Chi-square = 14.13; degree of freedom = 9; (0.100 < P < 0.250)

Table 2 HLA DQ Alpha Genotype Distribution in Black Population of Marion County, Indiana

HLA DQ Alpha GENOTYPE	OBSERVED (N = 203) ^a		EXPECTED (N = 202.6) ^a	
1.1, 1.1	3 ^b	(.015) ^c	5.7	(.028) ^c
1.1, 1.2	25	(.123)	21.0	(.104)
1.1, 1.3	1	(.005)	1.5	(.007)
1.1, 2	8	(.039)	6.0	(.023)
1.1, 3	7	(.034)	7.5	(.037)
1.1, 4	21	(.103)	20.3	(.100)
1.2, 1.2	19	(.094)	19.5	(.096)
1.2, 1.3	4	(.020)	2.8	(.014)
1.2, 2	12	(.059)	11.2	(.055)
1.2, 3	13	(.064)	14.0	(.069)
1.2, 4	34	(.168)	37.8	(.186)
1.3, 1.3	0	0	0.1	(.0004)
1.3, 2	0	0	0.8	(.004)
1.3, 3	3	(.015)	1.04	(.005)
1.3, 4	1	(.005)	2.7	(.013)
2, 2	2	(.010)	1.6	(.008)
2, 3	5	(.025)	4.0	(.020)
2, 4	7	(.034)	10.8	(.053)
3, 3	1	(.005)	2.5	(.012)
3, 4	15	(.074)	13.5	(.067)
4, 4	22	(.108)	18.3	(.090)

a: N = Number of people typed.

b: number of each genotype.

c: frequency of each genotype.

Chi-square = 5.51; degree of freedom = 7; (0.50 < P < 0.75)

Distribution of DQ α Allele in Marion County, FBI, and Cetus Populations

DQ α Allele	MARION COUNTY		FBI		CETUS	
	Caucasian N=185	Black N=203	Caucasian N=737	Black N=589	Caucasian N=826	Black N=448
1.1	0.124	0.167	0.131	0.134	0.137	0.150
1.2	0.238	0.310	0.205	0.270	0.197	0.263
1.3	0.068	0.022	0.075	0.042	0.085	0.045
2	0.127	0.089	0.138	0.126	0.109	0.121
3	0.176	0.111	0.182	0.113	0.201	0.118
4	0.268	0.300	0.282	0.322	0.271	0.304

Distribution of DQ Alpha Alleles in Marion County, FBI and Cetus Populations

