

Incidence of Rare Variants Among Serum Proteins and RBC Enzymes in US Whites and Blacks.

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The incidence of rare or uncommon genetic variants, including some population specific markers are difficult to assess due to small sample sizes generally reported. Such markers and the respective gene frequencies are of considerable value in parentage testing and as evidence in criminal investigations. For this study we have accumulated genetic data on the following marker systems markers GC, BF, F13A, F13B, HP, PLG, TF, ACPl, and PGMI from local white and black populations. Gene frequencies are presented using large sample sizes which provided data on 160 alleles, 32 new variants and 15 population specific markers.

#### METHODS

Phenotyping of genetic polymorphisms at our institution were performed using techniques previously published by the authors. Isoelectric focusing (IEF) was performed on all genetic marker systems except HP and BF, which were typed by conventional electrophoresis (CE). GC and PGMI were typed by IEF and CE. Unusual variants and rare variants were in many cases confirmed by the labs of Drs. P. Kuhl, G. Mauff, H. Cleve, S. Weidinger, J. Constans and Mr. M. Nelson.

#### RESULTS AND DISCUSSION

Tables 1-9 list the observed gene frequencies of the tested marker systems. The diagrams accompanying these tables are intended to demonstrate the position of the new variants which are marked with an asterisk (\*). For a more complete description of the new GC variants contact Dr. J. Constans, Toulouse, France. The number in closed parentheses ( ) to the right of the frequencies lists the number of observed families with this allele. All new variants had family data, but it may not have been listed because it was seen in another population group tested in our lab.

A descriptive summation of the results is not possible in this text, however table 10 gives an overview of the data. Of 160 alleles observed 135 were seen in whites and 70 in blacks. For alleles with frequencies of  $< 0.001$  we observed 104 in whites and 28 in blacks. Sample size may have contributed to this difference. For those alleles seen in both populations we classified them as population specific if they were different by a factor of  $\times 10$ , eg. PLG\*M1, ACPl\*R. Others, such as GC\*1C2, were called population specific if they were seen in only one population on a reoccurring basis.

This data gives a more reliable estimate of the frequency of rare and unusual alleles, because of the large populations tested. The data shows that there is a great deal of polymorphism within the protein and enzyme markers routinely used in many laboratories.

TABLE 1

		GC GENE FREQUENCIES	
		WHITE	BLACK
		(No. Tested 36,960)	(No. Tested 3,330)
1S	.....	0.56503	..... 0.18423
1F	.....	0.15526	..... 0.67222
2	.....	0.27802	..... 0.10706
1A1	.....	0.00004	..... 0.01712
1C10	.....	0.00001	..... 0.01622
1C1	.....	0.00035 (1)	..... -
1C2	.....	0.00024 (4)	..... -
1C3	.....	0.00026 (7)	..... 0.00045 (2)
1C6	.....	0.00014 (4)	..... -
1C12	.....	0.00005 (2)	..... -
1C13	.....	0.00003 (2)	..... -
1C16	.....	-	..... 0.00015
1C21	.....	0.00001	..... -
1C23	.....	-	..... 0.00015
1C25	.....	0.00001 (1)	..... -
1C31	.....	0.00005 (2)	..... -
1C32	.....	0.00005 (2)	..... -
1C33	.....	0.00004 (1)	..... -
*1C39	.....	0.00001	..... -
*1C40	.....	0.00001 (1)	..... -
*1C41	.....	0.00001	..... -
*1C48	.....	-	..... 0.00015
*1C51	.....	0.00001	..... -
*1C53	.....	-	..... 0.00015 (3)
2C7	.....	0.00001	..... -
2C11	.....	0.00001	..... -
*2C15	.....	0.00001 (1)	..... -
1A2	.....	0.00003 (1)	..... -
1A4	.....	0.00001 (1)	..... -
1A5	.....	-	..... 0.00015
1A7	.....	0.00001 (7)	..... -
1A8	.....	0.00005 (1)	..... -
1A10	.....	0.00008 (1)	..... -
1A11	.....	0.00001 (1)	..... -
1A12	.....	0.00001 (1)	..... -
1A19	.....	0.00001	..... -
*1A25	.....	-	..... 0.00015
1A29	.....	0.00001 (1)	..... -
2A2	.....	0.00005 (1)	..... -
2A3	.....	0.00001	..... 0.00135 (3)
2A5	.....	-	..... 0.00045 (1)
2A12	.....	0.00001	..... -
2A13	.....	0.00001 (1)	..... -
*2A16	.....	0.00001 (1)	..... -
*2A17	.....	0.00003	..... -

TABLE 2

	BF GENE FREQUENCIES	
	WHITE (No. Tested 41,657)	BLACK (No. Tested 3,564)
S	0.80238	0.44725
S07	0.00870	0.00828
F	0.17834	0.51207
F1	0.01024	0.03100
S030	0.00012 (4)	0.00014
S045	0.00010	0.00014 (1)
F025	0.00004 (1)	-
F030	0.00002	-
F055	0.00001	-
F075	-	0.00014 (1)
F080	0.00004	0.00098 (3)
F130	0.00001 (1)	-

TABLE 3

	F13A GENE FREQUENCIES	
	WHITE (No. Tested 2,152)	BLACK (No. Tested 255)
1	0.7664	0.792
2	0.2314	0.208
3	0.0002	-
4	0.0005 (1)	-
5	0.0002 (1)	-
*6	0.0002 (1)	-
*7	0.0005 (1)	-
*8	0.0002	-
*9	0.0002	-
*10	0.0002	-

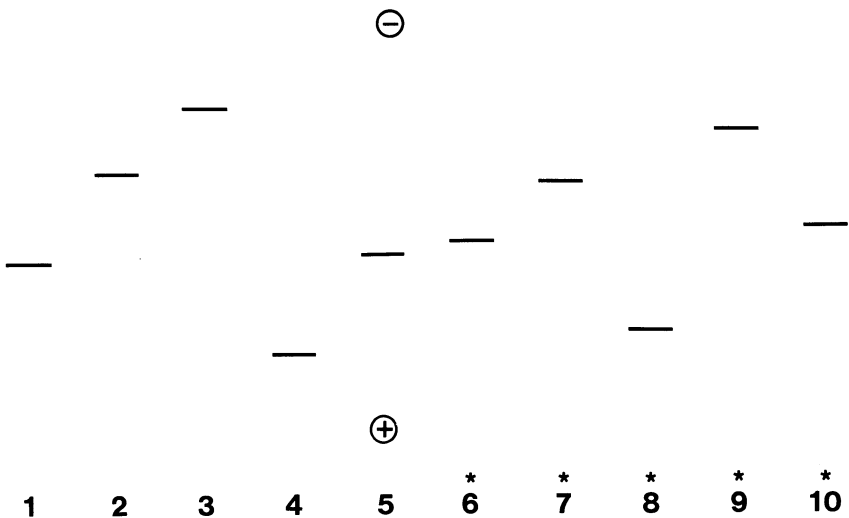


TABLE 4

		F13B GENE FREQUENCIES	
		WHITE	BLACK
		(No. Tested 22,344)	(No. Tested 2,380)
1	.....	0.74454	..... 0.34622
2	.....	0.09013	..... 0.56975
3	.....	0.16208	..... 0.06176
4	.....	0.00264	..... 0.00042
6	.....	0.00013	..... 0.01807
*16	.....	0.00013	..... -
*17	.....	-	..... 0.00084 (1)
*18	.....	-	..... 0.00252 (1)
*19	.....	0.00004 (1)	..... -
*20	.....	0.00027 (2)	..... -
*21	.....	0.00004 (1)	..... -
*22	.....	-	..... 0.00042 (1)

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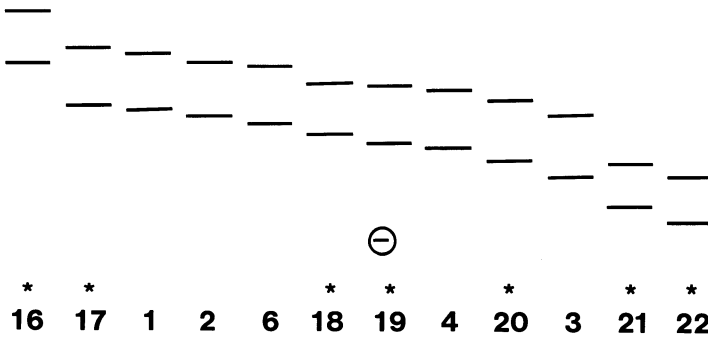


TABLE 5

		HP GENE FREQUENCIES	
		WHITE	BLACK
		(No. Tested 43,310)	(No. Tested 2,728)
1	.....	0.39651	..... 0.54384
2	.....	0.60254	..... 0.38979
B	.....	0.00002	..... -
CA	.....	0.00047	..... 0.00434
JO	.....	0.00021 (4)	..... 0.00041
MA	.....	0.00002 (1)	..... -
2MOD	.....	0.00023	..... 0.06162 (68)

TABLE 6

PLG GENE FREQUENCIES			
WHITE		BLACK	
(No. Tested 23,754)		(No. Tested 2,426)	
A	0.67888	0.71908	
B	0.28414	0.22464	
A3	0.01726	0.00412	
M1	0.00029	0.03936	
M5	0.01177	0.00350	
*A4	0.00042 (47)	0.00185	
A2	0.00002 (1)	-	
A1	0.00072	0.00041	
M4	0.00042	0.00021	
*M01	0.00352 (65)	0.00082 (1)	
*B01	0.00006 (3)	0.00041 (2)	
B1	0.00013	-	
B2	0.00002	0.00556	
*B21	0.00006	-	
*Q0	0.0035	-	

Untreated PLG

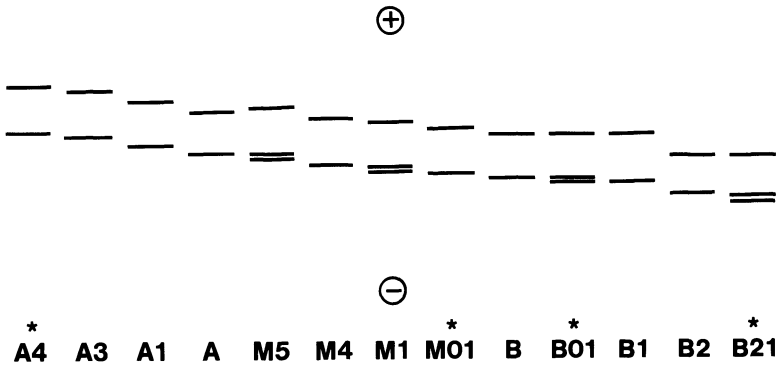


TABLE 7

TF GENE FREQUENCIES			
WHITE		BLACK	
(No. Tested 11,800)		(No. Tested 1,152)	
C1	0.78011	0.86330	
C2	0.15381	0.07856	
C3	0.05784	0.01432	
D1	0.00030	0.03863	
C4	0.00055	0.00043	
C5	0.00008 (2)	-	
C6	0.00047	-	
C7	0.00004	-	
C8	0.00008	0.00043 (1)	
C15	0.00004	0.00073 (1)	
B1	0.00004	-	
B0-1	0.00008	-	
B1-2	0.00131	0.00043	
B2	0.00470	0.00217	
D2	0.00017	-	
D3	0.00004	0.00043	
D4	0.00004	0.00007 (1)	

TABLE 8

	ACPI GENE FREQUENCIES	
	WHITE (No. Tested 43,424)	BLACK (No. Tested 3,560)
A	0.35086	0.22458
B	0.59075	0.74551
C	0.05824	0.01180
R	0.00003	0.01713
*A'	0.00006 (1)	0.00014 (1)
D	-	0.00014 (1)
E	-	0.00056 (2)
F	0.00001 (1)	-
*N	-	0.00014 (1)
*P	0.00001 (1)	-
*S	0.00001 (1)	-
*T	0.00002 (2)	-
*U	0.00001 (1)	-

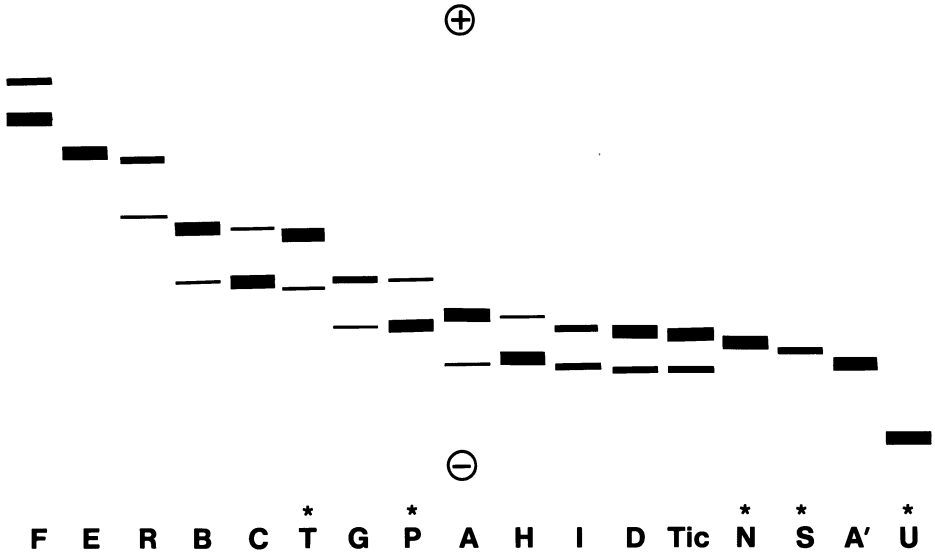


TABLE 9

		PGM1 GENE FREQUENCIES	
		WHITE	BLACK
		(No. Tested 38,930)	(No. Tested 3,492)
1A	.....	0.6355	..... 0.6591
1B	.....	0.1373	..... 0.1393
2A	.....	0.1700	..... 0.1617
2B	.....	0.0565	..... 0.0395
W1	.....	0.00001 (1)	..... 0.0001 (1)
W2	.....	0.00003 (2)	..... -
W3	.....	0.00010 (3)	..... -
W4	.....	0.00005 (1)	..... -
W5	.....	0.00001 (1)	..... -
W7	.....	0.00004 (2)	..... -
W9	.....	0.00005	..... -
W10	.....	0.00004 (3)	..... -
W11	.....	0.00003 (1)	..... -
W12	.....	0.00005 (3)	..... -
W14	.....	0.00003	..... -
W15	.....	0.00005 (3)	..... -
W16	.....	0.00001	..... -
W19	.....	0.00005 (3)	..... -
W21	.....	0.00003 (1)	..... -
W22	.....	0.00009 (2)	..... 0.0003 (1)
W26	.....	0.00001	..... -
W27	.....	0.00004 (1)	..... -
W30	.....	0.00001 (1)	..... -

TABLE 10

DISTRIBUTION OF ALLELES OBSERVED  
IN WHITES AND BLACKS

*System	# Alleles Observed	# Alleles 0.001	** New Variants	Population Specific
GC (48)			10	
W	38	35		1C1,1C2,1C6
B	14	9		1A1,1C10,2A3
BF (12)			-	
W	11	7		
B	8	3		F08
F13A (10)			5	
W	10	8		-
B	2	-		-
F13B (12)			7	
W	9	5		4
B	8	3		6
HP (7)			-	
W	7	5		-
B	5	3		2MOD
PLG (14)			4	
W	10	7		M01
B	8	4		B2,M1
TF (17)			-	
W	17	12		-
B	11	6		D1
ACP1 (13)			6	
W	10	7		-
B	8	4		R
PGM1 (27)			-	
W	23	18		-
B	6	2		-

\*Genetic marker tested. Number l ( ) is total number of variants observed. W = White, B = Black

\*\* Total number of new variants observed.