

## FREQUENCY OF FIVE GENETIC POLYMORPHISMS IN TWO POPULATIONS OF COLOMBIA

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### ABSTRACT

The prevalences of alleles from the glucose 6-phosphate dehydrogenase (G6PD), hemoglobin (Hb), ABO, Rh and haptoglobin (Hp) systems were investigated in two mixed Noanama Indian and Black Colombian populations. Among the Blacks the G6PD, Rh and Hp markers showed the expected frequencies, but the relatively high values found for *Hb\*C* (0.057) and *ABO\*O* (0.853), as well as the low one of *ABO\*B* (0.046) should be emphasized. The hemoglobin results can be explained by immigration of people from areas of high *Hb\*C* prevalences in Africa, or differential survival of carriers of this gene in Colombia, while the ABO findings may reflect Amerindian admixture. The presence of one G6PD-deficient subject, as well as the occurrence of *Hb\*C* (0.106), *ABO\*A* (0.260) and *ABO\*B* (0.027) among those labelled as predominantly Amerindian suggests heavy admixture, but the absence of Rh(-) among them is as expected. The number of haptoglobin typings performed in this group was low; the *Hp\*1* frequency observed (0.700) is similar to those found in South American Indians in general, but differs from the prevalence observed in a previous study of Noanama Indians.

### INTRODUCTION

The South American populations present an excellent opportunity for the study of microevolutionary phenomena. The variability of their founding stocks, and

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the preferential survival of their descendants when faced with different physical and socio-cultural environments gives the opportunity for a series of interesting insights about our biology. It is, therefore, regrettable that the amount of genetic information available for Colombian populations is still small. The present report is a modest attempt to start filling this gap.

## MATERIAL AND METHODS

Blood samples (5 ml) were taken from 191 male volunteers by venipuncture using ACD as anticoagulant, and refrigerated until analysis. They were obtained from 134 individuals of predominantly African origin from the localities of Guineal, Docordo and Orpua, and 57 predominantly Amerindians settled at Pichima and Union Balsalito of the Waunana (Noanama) linguistic group. These settlements are situated between the Baudó and San Juan Rivers (Chocó-Colombia).

Glucose-6-phosphate dehydrogenase activity was measured by fluorescence and spectrophotometry using Calbiochemical-Boehringer kits. Hemoglobin electrophoresis was carried out using 0.25 M Tris-EDTA borate buffer, pH 8.6, on cellulose acetate strips, according to the indication of Helena laboratories. Blood group systems were determined by standard hemagglutination procedures. Haptoglobins were measured by starch-gel electrophoresis using the same buffer as employed for the hemoglobins.

## RESULTS AND DISCUSSION

The localities and sample sizes are presented in Table I. Seventy percent of the individuals were of African origin. The distribution of G6PD activity among the predominantly Indian and Black populations is shown in Table II. G6PD-deficient individuals were classified as follows: the difference between the maximum value (14.470) and the minimum value (0.180), representing the range, was divided into ten

Table I - Localities and sample sizes.

Locality	Ethnic group	Number of samples	Total population
Guineal	Black	91	250
Orpua	Black	20	400
Pichima	Amerindian	26	340
Docordo	Black	23	750
Union	Amerindian	31	190

categories. Only individuals belonging to class 0 were considered deficient, and their frequency among Indians and Blacks was 1.8% and 12.7%, respectively.

Table II - Distribution of the G6PD activity in the predominantly Indian and Black populations.

Class	Range	Amerindians		Blacks	
		(No.)	(%)	(No.)	(%)
0	0.180 - 1.609	1	1.8	16	12.7
1	1.610 - 3.038	4	7.8	11	8.7
2	3.039 - 4.467	14	25.0	23	18.3
3	4.468 - 5.896	13	23.2	36	28.6
4	5.897 - 7.325	12	21.4	14	11.1
5	7.326 - 8.754	6	10.7	10	7.9
6	8.755 - 10.183	3	5.4	5	4.0
7	10.184 - 11.612	1	1.8	5	4.0
8	11.613 - 13.041	0	0.0	2	1.6
9	13.042 - 14.470	2	3.6	4	3.2
<b>Total</b>		<b>56</b>	<b>100.0</b>	<b>126</b>	<b>100.1</b>

The results concerning the four other systems are given in Table III. The following points are noteworthy: (a) Predominantly Black subjects: the G6PD, Rh and Hp alleles showed the expected prevalences but those of *Hb\*C* (0.057), and *ABO\*O* (0.853) are higher, while that of *ABO\*B* (0.046), is lower than expected. The hemoglobin results can be explained by immigration of people from areas of high *Hb\*C* in Africa, or differential survival of carriers of this gene in Colombia, while the ABO results may indicate Amerindian admixture. A previous study in a San Juan river population (Restrepo and Gutierrez, 1967) showed a prevalence of G6PD-negative subjects (13.5%) similar to that reported here. (b) Predominantly Amerindian individuals: The presence of one G6PD-deficient person, plus the frequencies of *Hb\*C* (0.106), *ABO\*A* (0.260) and *ABO\*B* (0.027) suggest a high level of miscegenation, but the absence of Rh negatives is as expected. Few subjects were tested for haptoglobin, but the *Hp\*1* value found is close to the South American Indian average. It should be mentioned, however, that Kirk *et al.* (1974) have previously sampled 168 Noanama Indians from this same region, observing absence of non-O or Rh(+) subjects, and a lower frequency of *Hp\*1* (0.432). The high *Hp\*1* prevalence found in our sample could be due to small sample size and/or to racial admixture.

Table III - Allele and phenotype frequencies for four genetic systems.

Genetic system and ethnic group	Observed phenotypes					Number studied	Allele frequencies		
	A	AC	AS	S	SC		<i>Hb*A</i>	<i>Hb*S</i>	<i>Hb*C</i>
Hemoglobin	A	AC	AS	S	SC		<i>Hb*A</i>	<i>Hb*S</i>	<i>Hb*C</i>
Blacks	99	10	1	1	3	114	0.917	0.026	0.057
Amerindians	41	11	0	0	0	52	0.894	0.000	0.106
ABO	A	B	AB	0			<i>ABO*O</i>	<i>ABO*A</i>	<i>ABO*B</i>
Blacks	21	7	5	101		134	0.853	0.101	0.046
Amerindians	24	1	2	30		57	0.713	0.260	0.027
Rh	Rh(+)	Rh(-)					<i>Rh*d</i>		
Blacks	117	3				120	0.158		
Amerindians	53	0.0				53	0.000		
Haptoglobin	1-1	2-1	2-2	0			<i>Hp*1</i>	<i>Hp*2</i>	
Blacks	40	38	13	24		115	0.648	0.352	
Amerindians	4	6	0	4		14	0.700	0.300	

As is also presented in Table III, a high percentage of the sample of African origin (20.9%) and of the predominantly Amerindian group (28.6%) presented anhaptoalbuminemia. This may be due to the frequent occurrence of hemolytic episodes among them. These episodes could have been caused by infection with malaria parasites, that are prevalent in this region, and/or to the administration of anti-malarial drugs to the G6PD-deficient subjects.

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### RESUMO

As prevalências dos alelos dos sistemas da glicose-6-fosfato desidrogenase (G6PD), hemoglobina (Hb), ABO, Rh e haptoglobina (Hp) foram investigadas em duas populações colombianas

miscigenadas de origem predominantemente indígena (da tribo Noanama) e africana. Entre os negróides os marcadores para G6PD, Rh e Hp mostraram as freqüências esperadas, mas os valores relativamente altos encontrados para *Hb\*C* (0,057) e *ABO\*O* (0,853), assim como o baixo para *ABO\*B* (0,046), devem ser enfatizados. Os resultados relativos à hemoglobina podem ser explicados por imigração de pessoas de áreas com altas prevalências de *Hb\*C* na África, ou sobrevivência diferencial de portadores deste gene na Colombia; enquanto os achados no ABO podem refletir mistura com Ameríndios. A presença de um indivíduo deficiente para G6PD, assim como a ocorrência de *Hb\*C* (0,106), *ABO\*A* (0,260) e *ABO\*B* (0,027) entre aqueles classificados como Ameríndios sugere nível alto de mistura, mas a ausência de Rh (-) entre eles está de acordo com o esperado. O número de tipagens para haptoglobina realizadas neste grupo foi baixo; a freqüência observada de *Hp\*1* (0,700) é similar à encontrada em índios sul-americanos em geral, mas difere da prevalência observada em um estudo prévio de índios Noanama.

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