THE DISTRIBUTION OF ENZYME GROUP SYSTEMS IN A SAMPLE OF SOUTH AFRICAN BANTU*

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A considerable number of enzyme systems are now known which show polymorphic variation in at least some human populations; the subject has been reviewed recently by Hopkinson[1] and Giblett.[2] The majority of studies have been carried out on persons of Caucasian origin, either in Europe or the USA, but an increasing number of reports are providing data on the variation in enzyme systems in other ethnic groups: among these are studies on black Africans or US Negroes.[3-17]

Much work still remains to be done before we have a complete picture of the world range of gene frequencies or an understanding of the factors responsible for the polymorphism in any of these systems. As a further contribution towards the completion of maps on the distribution of gene frequencies we have recently examined a series of Bantu in Durban for 8 red cell enzyme systems controlled by genes at 10 independent loci.

METHODS

Blood samples were collected by venepuncture from healthy Bantu blood donors attending the mobile clinic at the Natal Blood Transfusion Service. Portions of clotted blood (approximately 5·0 ml) were flown to Canberra in insulated containers within 48 hours of collection. On arrival the cells were separated, washed 3 times with normal saline and haemolysed by the addition of an equal volume of distilled water after the final wash. The haemolysates were stored at -20°C until used.

Starch gel electrophoretic conditions and methods for the visualization of enzyme patterns have been described in detail previously.[18,19]

RESULTS AND DISCUSSION

The number of persons tested and the distribution of types in each system is given in Table I. Appropriate χ^2 values, assuming the Hardy-Weinberg equilibrium, are also given in Table I. No significant departures from expectation occurred. Corresponding gene frequencies for systems where more than one allele is present are given in Table II.

Red Cell Acid Phosphatase (PHs)

Three alleles exist in many populations to control the phenotypic expression of red cell acid phosphatase: two of these, PHs^A and PHs^B are almost universally distributed. The third, PHs^C, appears to be absent or of very low frequency in black Africans, New Guineans and Australian Aborigines[1,20,21] as well as in Japanese.[22,23] In US Negroes and black Africans, however, a fourth allele, PHs^R, is present with reported frequency of 1 - 2%.[1,12]

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TABLE I. DISTRIBUTION OF RED CELL ENZYME GROUPS IN A SAMPLE OF SOUTH AFRICAN BANTU

System	*No.*	*Frequency*
Acid phosphatase		
A	4	01·32
AB	67	22·04
B	212	69·74
AR	4	01·32
BR	16	05·26
R	1	00·33
Total	304	
$\chi^2(3)$		1·9368
PGM_1		
1 - 1	219	72·04
2 - 1	82	26·97
2 - 2	3	00·99
Total	304	
$\chi^2(1)$		2·4357
PGM_2		
1 - 1	277	91·12
2 - 1	27	08·88
2 - 2	0	—
Total	304	
$\chi^2(1)$		0·6565
Peptidase A		
1 - 1	249	82·18
2 - 1	50	16·50
2 - 2	4	01·32
Total	303	
$\chi^2(1)$		0·6605
Peptidase B		
1 - 1	304	100·00
6-PGD		
AA	242	79·61
AC	58	19·08
CC	2	00·66
'Richmond'	1	00·33
'New'	1	00·33
Total	304	
$\chi^2(3)$		3·9259
AK		
1 - 1	298	98·03
2 - 1	6	01·97
2 - 2	0	—
Total	304	
$\chi^2(1)$		0·0302
LDH	304	100·00
MDH	304	100·00
'Oxidase'	304	100·00

TABLE II. GENE FREQUENCIES FOR RED CELL ENZYME GROUPS IN BANTU

Enzyme group	*Frequency*
Acid phosphatase	
PHs^A	0·1299
PHs^B	0·8339
PHs^R	0·0362
PGM_1	
PGM_1^1	0·8553
PGM_1^2	0·1447
PGM_2	
PGM_2^1	0·9556
PGM_2^2	0·0444
Peptidase A	
Pep A^1	0·9043
Pep A^2	0·0957
AK	
AK^1	0·9901
AK^2	0·0099
6-PGD	
PGD^A	0·8931
PGD^C	0.1036
$PGD^{Richmond}$	0·0033

The present survey confirms the absence of the PHs^C allele as well as the presence of the PHs^R in South African Bantu noted by Harris and his colleagues[1] though the present PHs^R frequency of 0·036 is slightly higher than the previously reported figure. In the present instance the PHs^R allele was present in 7% of the persons sampled and one person had a phenotype which has been interpreted as the homozygote expression of the PHs^R gene. The starch gel pattern of this phenotype is shown in Fig. 1. It is the first time that this phenotype has been demonstrated.

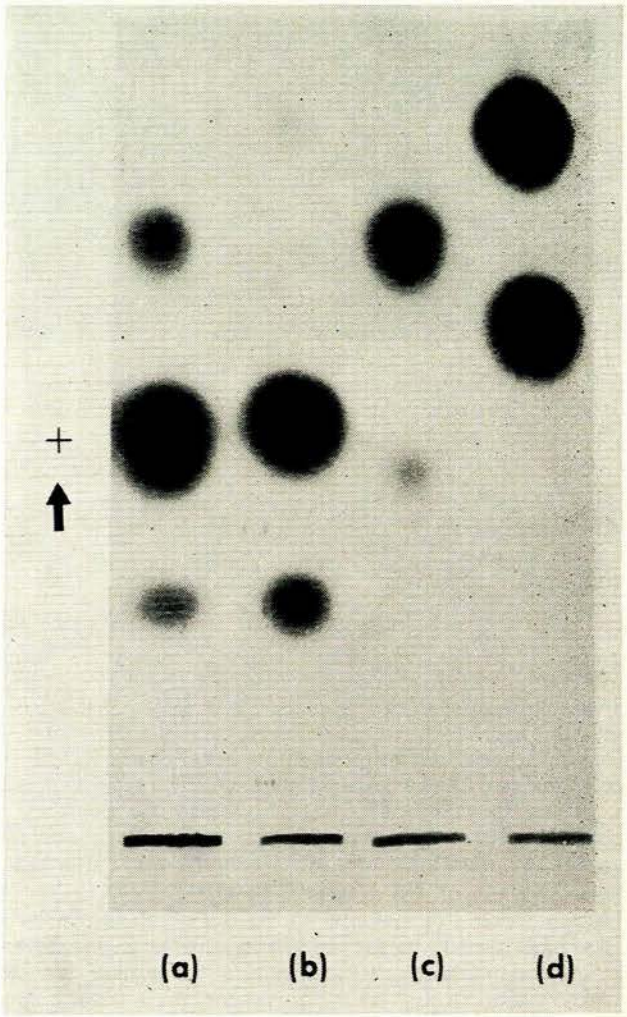

Fig. 1. Starch gel pattern of red cell acid phosphatase phenotypes: (*a*) AB, (*b*) B, (*c*) A, and (*d*) R. Phosphate/citrate/EDTA buffer pH 5·9.

Phosphoglucomutase (PGM)

Alleles at two independent loci PGM_1 and PGM_2 control the phenotypic expression of phosphoglucomutase activity in haemolysates of red cells; the activity of genes at a third locus, PGM_3, can be recognized in extracts of placentas or cultures of cells from other tissues.

At the PGM_1 locus the two common alleles, PGM_1^1 and PGM_1^2, are found in all populations, PGM_1^1 being

the commoner of the two. For Africans Hopkinson and Harris find PGM_1^1 frequencies of 0·76 for Yoruba in Nigeria and 0·79 for a sample of South African Bantu of unspecified origin. The value of 0·85 in the present study is not significantly different from the latter figure. No rare PGM_1 variants were found.

Variation due to alleles at the PGM_2 locus is of special interest because it is confined to US Negroes and black Africans. The first variant was described as the 'Atkinson' phenotype,[7] and this is the heterozygote combination PGM_2^1/PGM_2^2. Recently, a new phenotype at the PGM_2 locus has been described in a sample from pygmies in the Central African Republic.[16] This differs from the 'Atkinson' phenotype in the relative intensities of the *e* and *f* PGM isozymes.

In our present Bantu samples 8·9% gave patterns characteristic of the 'Atkinson' phenotype (Fig. 2). No cases were observed where band *e* was more intense than band *f* and we conclude that the $PGM_2^{'PYG'}$ allele is not present.

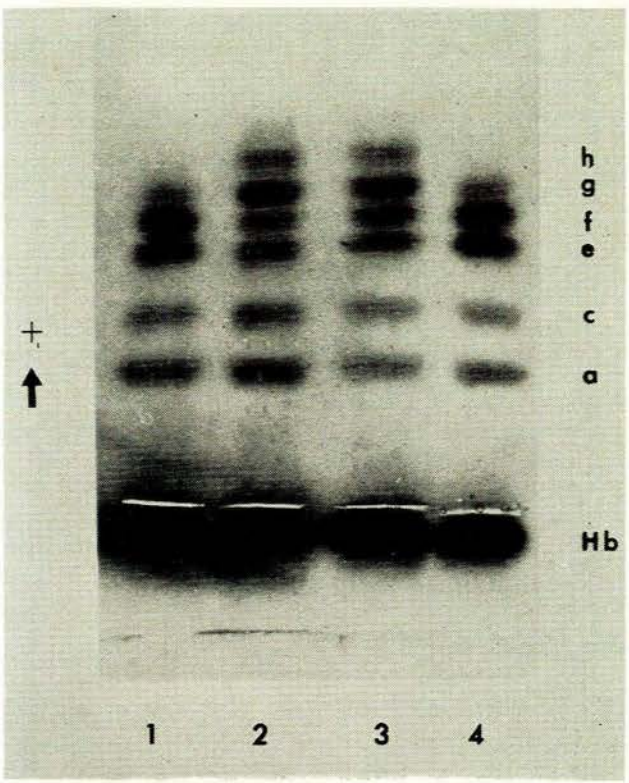

Fig. 2. Starch gel pattern of phosphoglucomutase locus 2 variants. Samples 1 and 4 PGM_2 1-1, samples 2 and 3 PGM_2 2 - 1. All samples are PGM_1 1 - 1. Tris/maleic acid/EDTA buffer pH 7·4.

The corresponding PGM_2^2 frequency is 0·04, a value considerably higher than that given by Hopkinson and Harris[1] for the Yoruba in Nigeria (0·006) and slightly higher than the same authors give for a small sample of South African Bantu (0·025).

Peptidases

The enzymes controlling peptidase activity are also under the control of genes at separate loci, and as in the case of the phosphoglucomutase system, alleles at one of these loci, Pep A, show polymorphic variation in Negroes but not elsewhere in the world.[24,25] Our present study provides the first figures for the Pep A system in South African Bantu. There were 16·5% Pep A 2 - 1 and 1·3% Pep A 2 - 2 phenotypes giving a frequency of 0·096 for the Pep A^2 allele. In one person the Pep A activity was too low to be typed.

Benerecetti[17] has described variation in activity of the Pep C system in pygmies which he considers to be due to a 'silent' allele, Pep C^0. We have not examined the Pep C system in our present series but for Pep B we found only the Pep B 1 - 1 phenotype, in agreement with the results of Lewis and Harris[24] for Negroes.

6-Phosphogluconate Dehydrogenase (6-PGD)

Two alleles in the 6-phosphogluconate dehydrogenase system, PGD^A and PGD^0, appear to be universally distributed[26] but the highest world frequency for PGD^C has been reported for South African Bantu sampled in Cape Town.[11] In addition to the common alleles other variants, such as 'Hackney', 'Richmond', 'Ilford' and 'Elcho' occur sporadically or with a highly localized distribution.[26,27]

Twenty per cent of the present series were variants of the normal 6-PGD pattern, 19·1% being the common 'Plaistow' variant (AC). Two persons with the 'Canning' phenotype (CC) were detected as well as one with the 'Richmond' phenotype. One individual had a hitherto undescribed phenotype. The fastest bands coincide in mobility with the anodal bands of the 'Richmond' phenotype, while the slowest bands coincide in mobility with the cathodal bands of the 'Plaistow' or 'Canning' variants. This person is considered to be the heterozygote $PGD^C/PGD^{RICHMOND}$; unfortunately the father of the propositus is deceased but her mother has the 'Richmond' phenotype and her sib is normal. The over-all PGD^C frequency of 0·104 is lower than that reported by Gordon *et al.*[11] for Bantu in Cape Town, but it is still among the highest world frequencies for this allele.

Adenylate Kinase (AK)

The three phenotypes, AK 1 - 1, 2 - 1 and 2 - 2 are controlled by two codominant alleles, AK^1 and AK^2. The AK^2 gene has a frequency approximating 0·05 in European populations but is rarer in US Negroes and black Africans[1] and is absent in New Guineans and Australian Aborigines.[20,21,28] The highest frequencies for AK^2 occur in India.[19,29] Six AK 2 - 1 persons were found in the present survey, giving an AK^2 frequency of 0·01. This figure is identical with that for a series of 100 South African Bantu studied by Rapley *et al.*[14]

Lactate Dehydrogenase, Malate Dehydrogenase and 'Oxidase'

The 3 remaining enzyme systems revealed no variation from the normal pattern among the 304 Bantu blood specimens examined in the present survey.

LDH variants have been reported sporadically from many populations[30-32] and in US Negroes the frequency of variants approximates 1%.[30,33] In India even higher frequencies ranging up to 4% have been reported.[34] Because of the reported incidence of LDH variants in US Negroes and in a small sample of 23 persons from

Nigeria it is surprising that no variants were encountered here. For a low frequency event, however, this may have been due to chance and further investigation of a larger number of samples is required.

Variants of soluble malate dehydrogenase are extremely rare, only one case having been published so far,[35] although recently several examples of another S-MDH variant have been found in samples from New Guinea.[36] Because of the rarity of MDH variants it is not surprising that no examples were found in the present survey. Similarly, variation in the 'achromatic' zones detectable on tetrazolium-stained gels is also a great rarity. These zones have been identified with 'oxidase' activity.[37]

SUMMARY

The distribution of electrophoretic variants in 8 red cell enzyme systems representing 10 independent gene loci has been examined in 304 blood samples from South African Bantu in Natal.

In the acid phosphatase system no phenotypes containing the C components were detected, but 7% of samples contained an R component, the PHs^R frequency being 0·036. One individual was homozygous PHs^R/PHs^R. For the PGM_1 locus the PGM_1^1 frequency was 0·855 and no uncommon variants were detected. Nearly 9% of the persons tested showed variation at the PGM_2 locus, the PGM_2^2 frequency being 0·044.

No peptidase B variants were detected but the frequency of Pep A^2 was 0·096. Similarly, the PGD^0 allele had a relatively high frequency (0·103) and one 'Richmond' phenotype and one presumed heterozygote $PGD^0/PGD^{Richmond}$ were detected in the 6-phosphogluconate dehydrogenase system.

Few variants in the adenylate kinase system were present, the AK^2 frequency being 0·010. No variants for LDH, soluble MDH or 'oxidase' were detected.

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