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Further population data on red cell acid phosphatase, phosphoglucomutase and adenylatekinase polymorphisms in Sardinia

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Genetica. — *Further population data on red cell acid phosphatase, phosphoglucomutase and adenylatekinase polymorphisms in Sardinia* (*). Nota (**) di SILVANA A. SANTACHIARA-BENERECETTI (***), FLAMINIO BRUNELLI (****), FRANCA GIGLIANI (****), BACHISIO LATTE (*****), GUIDO MODIANO (****), MARIO NEGRI (***), CARLO SANTOLAMAZZA (****), ROSARIA SCOZZARI (****) e LUCIANO TERRENATO (****), presentata dal Socio G. MONTALENTI.

RIASSUNTO. — Sono state determinate le frequenze geniche per la A.P., per la PGM e per la AK eritrocitarie in Sardegna. I nuovi dati confermano quelli raccolti precedentemente. In particolare la frequenza dell'allele PGM_1^2 è significativamente, anche se di poco, inferiore a quella osservata nella popolazione di Roma; la frequenza dell'allele AK^2 è la più bassa finora osservata in una popolazione caucasica, mentre le frequenze geniche della A.P. sono uguali a quelle trovate a Roma.

The data reported in the present paper are part of a long term population study performed by our group during recent years to investigate the relationship, if any, between malaria and the polymorphisms for red cell acid phosphatase (A.P.), red cell phosphoglucomutase (PGM_1 and PGM_2) and red cell adenylate kinase (AK).

The available data on these polymorphisms have been extensively reviewed by Giblett in 1969 [1].

The last reports on A.P. [2] and on PGM and AK in Sardinia [3] have been summarized [4]. Since then, we have collected more data while trying to describe the biochemical make-up of red cells from normal, thalassemic and Glucose-6-Phosphate dehydrogenase deficient individuals.

These results will be the subject of further publications.

In the meanwhile we consider worthwhile to give an up-to-date account of the population data.

THE SAMPLES.

The subjects (284 for A.P., 288 for PGM and 294 for AK) on whom the present investigations were carried out, as well as their parents, were born in Sardinia. They were unrelated adult male individuals randomly selected among students of University of Cagliari, patients of Psychiatric Hospital

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of Cagliari, workers of the Centro Antiinsetti, persons doing their military service and relatives of patients with Cooley's anemia.

Since we have found no indication of heterogeneity of the distribution of these genes in Sardinia [2, 3] nor any discrepancy between the already published data and the present ones, we report all the data pooled together.

METHODS.

The bloods were collected by vein puncture with heparin vacutainers (Beckton, Dickinson and Company – Columbus, Nebraska) and kept cool (4° C to 10° C) until used.

The hemolysates were obtained by a duplicated cycle of freezing and thawing of the packed and twice washed red cells. A mixture of acetone and dry ice was used for the freezing and a 37° C water-bath for the thawing.

The electrophoretic phenotypes were determined by micromethods [5] derived from the standard techniques for A.P. [6] with minor modifications [2]; for PGM [7] and for AK [8].

RESULTS AND DISCUSSION.

The distributions of the phenotypes of these three genetic polymorphisms are shown in the Tables I, II and III.

TABLE I.

Distribution of red cell acid phosphatase phenotypes in a sample of 637 (*) Sardinians.

Phenotypes	Incidence (in percent)	Absolute frequencies		Chi-square (Yates)
		Observed	Expected	
A	6.0	38	41.7	0.247
BA	34.2	218	213.7	0.069
B	42.7	272	273.6	0.005
CA	5.0	32	28.9	0.230
CB	11.5	73	74.1	0.004
C	0.6	4	5.0	0.052
Totals	100.0	637	637.0	0.607

Chi-square (3 d.f.) = 0.607, P > 0.80
(for deviation from Hardy-Weinberg equilibrium)

Estimates of gene frequencies:
$P^a = 25.6 \pm 1.22$
$P^b = 65.5 \pm 1.33$
$P^c = 8.9 \pm 0.80$
100.0

(*) This figure is comprehensive of the sample of 365 individuals previously published [2]. Among the 284 new subjects, 12 have been excluded because no unambiguous classification was attained.

TABLE II.

Distribution of red cell phosphoglucomutase phenotypes in a sample of 921 (*) *Sardinians.*

Phenotypes	Incidence (in percent)	Absolute frequencies		Chi–square
		Observed	Expected	
1–1	58.9	542	541.81	0.000
2–1	35.6	328	329.18	0.004
2–2	5.5	51	49.99	0.020
Totals	100.0	921	920.98	0.024

Chi–square (1 d.f.) = 0.024 $P > 0.80$
(for deviation from Hardy–Weinberg equilibrium)

Estimates of gene frequencies: $PGM_1^1 = 76.7 \pm 0.98$; $PGM_1^2 = 23.3$; 100.0

(*) This figure is comprehensive of the sample of 633 individuals previously published [3].

TABLE III.

Distribution of red cell adenylate kinase phenotypes in a sample of 1327 (*) *Sardinians.*

Phenotypes	Frequencies	
	Relative	Absolute
1–1	97.6	1295
2–1	2.3	31
2–2	0.1	1
Totals	100.0	1327

Estimates of gene frequencies: $AK_1 = 98.8$; $AK_2 = 1.2 \pm 0.21$; 100.0

(*) This figure is comprehensive of the sample of 1033 individuals previously published [3].

These new data confirm the previous findings.

In fact the present A.P. gene frequencies are the same as those found in Rome [2]; on the other hand the gene frequencies of the other two polymorphisms are different: the PGM_1^2 and AK^2 gene frequencies are in Rome 0.287 [9] and 0.037 [10] respectively, while in Sardinia are 0.233 (see Table II) and only 0.012 (see Table III). The Chi-squares (1 d.f.) for the comparisons of these two populations are 8.28 with $P < 0.005$ for PGM_1, and 29.77 with $P << 0.0001$ for AK.

As a matter of fact this AK^2 gene frequency, besides being the lowest observed among Caucasians, is in the range observed among American Negroes, who present this gene only by genetic admixture [1].

The Sardinian population continues to show the uniqueness of its genetic make-up, which involves not only the *Gd* and *Th* genes, but also many other genes, whose distributions are not affected by malaria (ABO, MN, Rh) [11].

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