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Genetica. — *Distribution of red cell adenosine deaminase, phosphoglucomutase, and esterase-D isozymes in southern Latium.* Nota di GIORGIO BATTISTUZZI (*), CARMEN CASSERO, PAOLA IUDICONE, ROMANO PETRUCCI, P. SANTOLAMAZZA, ROSARIA SCOZZARI e MAURO SERAFINI, presentata (**) dal Socio G. MONTALENTI.

RIASSUNTO. — Sono stati studiati 401 soggetti di Fondi, Gaeta e Formia (località della costa meridionale del Lazio) per gli enzimi eritrocitari ADA, PGM_1 e ESD. I valori di frequenze geniche trovati in questi campioni sono simili a quelli precedentemente descritti per Roma e Lazio. Nel caso della ESD è stata trovata una notevole eterogeneità tra i tre campioni, nonostante la loro vicinanza geografica, da attribuirsi probabilmente a fluttuazioni casuali delle frequenze geniche.

Available data on the distribution of polymorphic enzyme allele frequencies have been collected since 1962 in Italy. In spite of this, the data are fully informative only for Sardinia, the population of which has aroused the interest of population geneticists for a number of reasons such as the pattern of adaptation to malaria and isolation. On the other hand, data concerning the pattern of erythrocyte genetic polymorphisms in the population of the peninsula are rather sporadic with respect to both the geographic distribution and the number of analyzed markers. The present paper is part of a systematic study of the genetic structure of the Italian population. We report here the distribution of red cell adenosine deaminase (ADA), phosphoglucomutase, locus 1 (PGM_1) and Esterase D (ESD) in Southern Latium.

MATERIALS AND METHODS

Blood samples were withdrawn by venipuncture into heparinized test tubes from 13-14 year old male and female subjects unrelated and apparently healthy, living in Southern Latium: 138 individuals from Fondi, 118 from Gaeta and 145 from Formia. Erythrocytes were washed three times with cold isotonic saline and lysed by repeated freezing and thawing.

ADA, PGM and ESD electrophoretic phenotypes were determined according to the method of Spencer *et al.*, 1968; Spencer *et al.*, 1964 and Hopkison *et al.*, 1973, respectively.

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RESULTS AND DISCUSSION

Phenotype and gene frequencies for the three markers are shown in Table I. A close correspondence between observed and expected frequencies according to Hardy-Weinberg equilibrium is verified in each case.

TABLE I

Phenotype and allele frequencies of the polymorphic genes ADA, PGM_1 and ESD *in Southern Latium.*

Sample	No.	Phenotypes (1)			Allele frequencies (2)		SE (2)	χ^2_1 (3)
		1	2 1	2	*p*	*q*		
ADA								
Fondi . .	136	119 *117.64*	15 *17.68*	2 *0.68*	93.0	7.0	1.60	2.984
Gaeta . . .	113	94 *94.81*	19 *17.39*	0 *0.80*	91.6	8.4	1.84	0.956
Formia . .	145	123 *122.99*	21 *21.10*	1 *0.90*	92.1	7.9	1.58	0.012
PGM_1								
Fondi . .	133	70 *71.42*	55 *52.00*	8 *9.44*	73.3	26.7	2.71	0.421
Gaeta . . .	117	54 *55.38*	53 *50.23*	10 *11.39*	68.8	31.2	3.03	0.357
Formia . .	145	67 *68.23*	65 *62.47*	13 *14.30*	68.6	31.4	2.72	0.243
ESD								
Fondi . .	138	110 *109.57*	26 *26.77*	2 *1.66*	89.1	10.9	1.86	0.093
Gaeta . . .	118	86 *85.66*	29 *29.76*	3 *2.58*	85.2	14.8	2.31	0.089
Formia . .	145	95 *95.13*	45 *44.63*	5 *5.23*	81.0	19.0	2.30	0.013

(1) Expected numbers in italics.
(2) In percent.
(3) χ^2 values for consistency with Hardy-Weinberg formula.

Heterogeneity in the distribution of alleles between isolates is statistically significant only for Esterase D ($\chi^2_2 = 6.70$, $0.05 > P > 0.02$).

This unequal distribution reflects the great variability of *ESD* allele frequencies in different Italian groups, ESD^2 ranging from 0.05 to 0.19 (see Battistuzzi *et al.*, 1978).

As far as ADA and PGM_1 are concerned the allele frequencies found in our samples do not significantly deviate from those found for the population

of Rome (Modiano *et al.*, 1970; Scozzari *et al.*, 1970; Battistuzzi *et al.*, 1974) or for the other continental Italian groups so far studied (see Scozzari *et al.*, 1978, and Battistuzzi *et al.*, 1978).

REFERENCES

HOPKINSON D. A., MESTRINER M. A., CORTNER J. and HARRIS H. (1973) – *Esterase D: a new human polymorphism*, « Ann. Hum. Genet., Lond. », *37*, 119.

MODIANO G., SCOZZARI R., GIGLIANI F., SANTOLAMAZZA C., AFELTRA P. and FRATTAROLI W. (1970) – *Red cell phosphoglucomutase polymorphism.* i. *Enzyme activity of different red cell* PGM *phenotypes*, « Hum. Hered. », *20*, 83.

SCOZZARI R., LUCARINI N. and DISCEPOLI L. (1978) – *Red cell adenosine deaminase polymorphism in the Marca di Camerino (Central Italy)*, « Rend. Accad. Naz. Lincei », in press.

SCOZZARI R., SANTOLAMAZZA C. and CARAPELLA E. (1970) – *Studies of the red cell adenosine deaminase polymorphism in Rome*, « Humangenetik », *8*, 364.

SPENCER N., HOPKINSON D. A. and HARRIS H. (1964) – *Phosphoglucomutase polymorphism in man*, « Nature », *204*, 742.

SPENCER N., HOPKINSON D. A. and HARRIS H. (1968) – *Adenosine deaminase polymorphism in man*, « Ann. Hum. Genet. », London,m *32*, 9.

BATTISTUZZI G., SANTOLAMAZZA C., MASTRANTONIO M., IUDICONE P., URBANI F., CAIOLA S., SANTOLAMAZZA P. and MANUALI A. R. – *Erythrocyte enzyme polymorphisms in Italy*, in preparation.

BATTISTUZZI G., SCOZZARI R., SANTOLAMAZZA P., TERRENATO L. and MODIANO G. (1974) – *Comparative activity of red cell adenosine deaminase allelic forms*, « Nature », *251*, 711.