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Red cell adenosine deaminase polymorphism in the Marca di Camerino (Central Italy)

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Genetica. — *Red cell adenosine deaminase polymorphism in the* Marca di Camerino (*Central Italy*). Nota di ROSARIA SCOZZARI (*), NAZZARENO LUCARINI (**) e LORETTA DISCEPOLI (**), presentata (***) dal Socio G. MONTALENTI.

RIASSUNTO. — Sono state determinate le frequenze geniche per la adenosindeaminasi (ADA) eritrocitaria umana in un campione di 278 individui originari della zona denominata *Marca* di Camerino (Italia Centrale). La frequenza dell'allele ADA^2 (.070 ± .011) è molto simile a quella trovata nella zona del delta del Po e non differisce in modo significativo da quelle osservate nelle altre popolazioni dell'Italia Continentale studiate finora.

INTRODUCTION

It is now clear that biochemical genetic polymorphisms, besides showing a high degree of heterogeneity among large racial groups, exhibit also a wide variability even among smaller groups [1].

The main alternative to selection as the cause of the diversity often observed between different groups is genetic drift. This is more likely to occur if the population is small and relatively isolated.

One such population is that living in the area named *Marca di Camerino* (Central Italy) (fig. 1). This region arose under the rule of Charles the Great as frontier land. It includes 24 mountain villages, difficult to reach, having a number of inhabitants ranging from a few hundred to a few thousand. These villages, as a whole, have been isolated—at least culturally—for centuries from neighbouring populations as documented by the persistence of an economy based almost exclusively on crops and sheep-rearing. Even for the present generation the degree of genetic isolation of this population is very high: more than 80 per cent of individuals have both parents born in the same village.

The present paper reports the adenosine deaminase (ADA) phenotype and gene frequencies in a random sample of 278 unrelated individuals from the *Marca di Camerino* area.

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(***) Nella seduta del 14 gennaio 1978.

Fig. 1. – Map of Italy showing the *Marca di Camerino* area.

SAMPLE AND METHODS

Unrelated individuals born in the *Marca di Camerino* area, as were their parents, have been examined. These were adult individuals of both sexes randomly selected through the Transfusion Centre or through the Civil Hospital of Camerino (they were affected by no hematological diseases).

Blood specimens, collected in .04 M sodium citrate, .02 M citric acid and .14 M dextrose (ACD) solution, were kept refrigerated and tested within three days from the time of collection.

Packed red cells washed three times with isotonic saline were lysed by a triplicate cycle of freezing and thawing. Soon after horizontal starch gel electrophoresis was carried out according to the method of Spencer *et al.*, 1968 [2].

TABLE I

Distribution of red cell adenosine deaminase phenotypes in a sample of 278 individuals from the Camerino area.

Phenotypes	Incidence	Absolute frequencies	
		Observed	Expected
ADA 1	86.0	239	240.44
ADA 1–2	14.0	39	36.20
ADA 2	0.0	0	1.36
Totals . . .	100.0	278	278.00

Estimates of gene frequencies

$ADA^1 = 0.93$

$ADA^2 = 0.07 \pm 0.011$

1.00

TABLE II

Red cell adenosine deaminase gene frequencies in various continental Italian populations.

Population	No. tested	ADA^2 gene frequency	χ^2_{1df}	P	References
Marca di Camerino . .	278	.070	—	—	This paper
Po Delta.	487	.074	.08	>.90	[3]
Bologna	276	.091	1.57	>.20	[4]
Padua	410	.080	.50	>.50	[5]
Pisa and Leghorn . .	135	.052	1.01	>.30	[6]
Rome	320	.089	1.44	>.20	[7]
Rome	196	.080	.68	>.50	[8]

RESULTS AND DISCUSSION

The phenotype and the gene frequency estimates are given in Table I. Only two out of the three red cell ADA phenotypes, first described by Spencer *et al.*, 1968 [2], have been found in the present sample, therefore the Hardy-Weinberg equilibrium was not testable.

The ADA^2 gene frequency found in the present sample is $.070 \pm .011$; this frequency is remarkably similar to that observed in the region of the Po Delta [3].

As far as comparisons with other continental Italian populations are concerned, no significant differences emerged between the present population and the other populations studied from this viewpoint (see Table II). Moreover, the ADA^2 gene frequency distribution appears to be homogeneous among the continental Italian areas so far examined, as was evaluated by a χ^2 test for heterogeneity ($\chi^2_{6df} = 5.23$; $P > .50$).

As these data show, there is no evidence to support the idea that genetic drift may have been an important agent in determining the genetic makeup of the *Marca di Camerino* population, at least as far as the ADA polymorphism is concerned.

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