THE RED CELL ACID PHOSPHATASE POLYMORPHISM IN GREEK b-THALASSEMIA PATIENTS

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Red cell acid phosphatase polymorphism was studied by starch gel electrophoresis in 70 b-thalassemia patients and in 310 healthy Greeks. Our results gave the following gene frequencies: b-thalassemia patients: p^a 0.321, p^b 0.643, p^c 0.036; healthy Greeks: p^a 0.302, p^b 0.653, p^c 0.045. No statistically significant differences were found between the two groups.

Red cell acid phosphatase (AP) polymorphism has been first described by Hopkinson et al. (1963). It appears as though this polymorphism is determined by a series of codominant alleles on a single locus. Three of these alleles p^a, p^b and p^c, are very common, while the other two, p^r and p^d, are rarely observed (Giblett 1969). Several studies failed to show any correlation between red cell AP and other polymorphic systems (Conneally et al. 1965, Schlesinger and Sulich 1970). We have now examined red cell AP in b-thalassemia patients as compared to healthy Greeks.

ACD blood samples from unrelated 310 apparently healthy donors and 70 b-thalassemia patients were collected. The patients had been transfused at least two months before the venipuncture. The AP phenotypes were determined by the horizontal starch gel electrophoresis according to Hopkinson et al. (1964) with the following modifications: Bridge buffer, citrate-phosphate buffer (NaH_2PO_4 0.245M — $Na_3C_6H_5O_7$ 0.15M); pH, 5.9. Gel buffer was prepared by diluting the bridge buffer 1 : 100 in distilled water. Statistical analysis was performed by using the Brand and Snedecor formula (Bailey 1969).

Our results are shown in the Table.

The p^b gene is the most frequent among the different races, but its frequency in whites is usually lower than 0.70, while in Negroes it is higher than 0.75 (Hopkinson and Harris 1968). On the other hand, the BA phenotype is the most frequent among whites, while the B phenotype is most frequent among Negroes and Orientals (Schlesinger and Sulich 1970). In our material the frequencies of p^b gene and BA phenotype were found as expected in whites.

Previous studies concerning red cell AP polymorphism in healthy Greeks were performed by Kattamis et al. (1965) and Stamatoyannopoulos et al. (1975). Whereas no significant differences appear between the two samples in our material, the AP phenotypic distribution in our b-thalassemia patients differs significantly from that found in healthy Greeks by Kattamis et al. (1965) and Stamatoyannopoulos et al. (1975). On the other hand, gene frequencies in our material are in good agreement with those estimated by Stamatoyannopoulos et al. (1975), but are significantly different from those found by Kattamis et al. (1965). The small sample and perhaps the nonrandom-mating population tested could explain this discrepancy.

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Table. *Red cell acid phosphatase in healthy and b-thalassemic Greeks*

A. *Results of Present Study*

Phenotypes	Healthy		b-thalassemic	
	No.	%	No.	%
A	18	5.81	2	2.86
B	126	40.65	22	31.43
C	1	0.32	0	0
BA	139	44.84	41	58.57
CA	12	3.87	0	0
CB	14	4.51	5	7.14
Total	310	100.00	70	100.00
Gene frequencies:	p^a 0.302, p^b 0.653, p^c 0.045		p^a 0.321, p^b 0.643, p^c 0.036	

B. *Comparison with Previous Studies: Phenotypes*

	B	BA	Others	Total	χ^2 test	
1. b-thalassemia Greeks	22	41	7	70	1 vs. 2a: 4.4	$p > 0.1$
2. Healthy Greeks					1 vs. 2b: 8.0	$p < 0.025$
a. Present study	126	139	45	310	1 vs. 2c: 6.58	$p < 0.05$
b. Stamatoyannopoulos et al. (1975)	244	250	117	611	2a vs. 2b: 3.36	$p > 0.1$
c. Kattamis et al. (1965)	45	37	14	96	2a vs. 2c: 1.33	$p > 0.5$

C. *Comparison with Previous Groups: Gene Frequencies*

	p^a	p^b	p^c and others	Total	χ^2 test	
1. b-thalassemia Greeks	0.321	0.643	0.036	1.000	1 vs. 2a: 1.64	$p > 0.4$
2. Healthy Greeks					1 vs. 2b: 1.08	$p > 0.5$
a. Present study	0.302	0.653	0.045	1.000	1 vs. 2c: 33.24	$p < 0.000007$
b. Stamatoyannopoulos et al. (1975)	0.322	0.633	0.045	1.000	2a vs. 2b: 0.96	$p > 0.95$
c. Kattamis et al. (1965)	0.218	0.714	0.068	1.000	2a vs. 2c: 20.96	$p < 0.0005$

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