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Red Cell Esterase-D-Polymorphism in the Veneto Population

P. Cortivo[1], F. Breda[1], G. Ongaro[2], G. Bareggi[2] and L. Ferraretto[2]

[1] Istituto di Medicina legale e delle Assicurazioni dell'Università di Padova, Via Faloppio 50, I-35100 Padova, Italy
[2] Servizio di immunoematologia e trasfusionale dello Ospedale Civile di Padova

Summary. 630 individuals and 102 mother/child pairs from the Veneto provinces were examined for their red cell EsD by Cellogel electrophoresis. Compared with the horizontal starch-gel electrophoresis no differences in the zymogram patterns have been found.
The gene frequencies observed (EsD^1 = 0.8556; EsD^2 = 0.1444) differ only slightly from those reported for various European populations and are nearly identical with data for other Italian populations.
Family studies confirmed the Mendelian co-dominant inheritance.

Key words: Blood Groups, Esterase-D – Esterase-D-Polymorphism

Zusammenfassung. 630 Einzelpersonen sowie 102 Mutter/Kind-Paare aus den Veneto Provinzen wurden zur Bestimmung ihrer Erythrocyten EsD mittels Cellogel-Elektrophorese untersucht. Im Vergleich zur horizontalen Stärkgel-Elektrophorese wurden bei den Zymogramm-Mustern keine Unterschiede entdeckt. Die beobachteten Genfrequenzen (EsD^1 = 0,8556; EsD^2 = 0,1444) unterscheiden sich nur geringfügig von den anderer europäischer Bevölkerungsgruppen und sind beinahe identisch mit Daten über andere italienische Bevölkerungsgruppen.
Familienstudien bestätigten die mendelische co-dominante Vererbung.

Schlüsselwörter: Blutgruppen, Esterase-D – Esterase-D-Polymorphismus

Esterase-D is an enzyme present in all human tissues of hydrolizing 4-methyl-umbelliferyl-acetate and -butyrate. Hopkinson et al. [1] demonstrated the enzyme polymorphism and described three phenotypes EsD1, EsD1–2 and EsD2 determined by two co-dominant, autosomic alleles EsD^1, EsD^2. Rare variants, EsD3–1, EsD4–1 and EsD4–2 have also been found [2, 3, 4, 5]. In the present work the EsD gene frequency in a Veneto population has been investigated.

Materials and Methods

The genetic frequency investigation has been performed on a sample of 630 subjects, from the Veneto provinces. Furthermore, blood samples of mothers and of the umbilical cord of their new-born child were examined.

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Table 1. EsD phenotype distribution and gene frequences in the population of Veneto

EsD	Phenotypes				χ^2
	observed		expected		
	n	%	n	%	
EsD1	457	72.6	461.2	73.20	0.0382
EsD2–1	164	26.0	155.67	24.71	0.4457
EsD2	9	1.4	13.13	2.09	1.2990
Total	630	100.00	630.00	100.00	1.7829

$\chi^2 = 1.7829$ for 1 df $0.2 > P > 0.1$
Gene frquencies: $EsD^1 = 0.8556$; $EsD^2 = 0.1444$

Table 2. Mother/child combinations in EsD polymorphism

Mothers	Children			
	EsD1	EsD2–1	EsD2	n
EsD1	58	14	-	72
EsD2–1	14	15	1	30
EsD2	-	-	-	-
n	72	29	1	102

Electrophoresis of hemolysates was performed on Cellogel sheets at 180 V for 1^h 45 min at room temperature; Bridge buffer: phosphate – citrate pH 6.4

Demonstration of the EsD bands (for 5.7 x 11 Cellogel sheets): 100 mg agar were dissolved in 7 ml acetate buffer pH 5.2; 10 mg 4-methylumbelliferyl-acetate were dissolved in 3 ml acetone and mixed with the agar solution at 50°C, which was layered on a 5.7 x 11 cm glass plate. After the electrophoretic separation the Cellogel sheets were laid upside down on the surface of the agar gel. After incubation for 10 min at 37°C in wet chamber the EsD bands were visible as fluorescent patterns under U. V. light (366 um).

Control samples were investigated by the horizontal starch-gel electrophoresis technique described by Hopkinson et al. [1].

Results and Discussion

The zymogram patterns of the EsD phenotypes obtained by electrophoresis on Cellogel were identical with those obtained by starch-gel electrophoresis. However, using the second method the bands were sharper.

The distribution of the phenotypes in the population sample is shown in Tab. 1. The χ^2 value follows the Hardy-Weinberg law. The gene frequencies observed differ only slightly from those found by other authors for various European populations (Ebeli-Struijk [6] et al. and Blake [7]) are nearly identical with those of the Tuscan (3) and Ligurian (8) populations, the only samples examined in Italy.

The results of mother/child pairings are reported in Tab. 2. No „impossible" mother/child combination occurred confirming the co-dominant heredity.

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