Genetic polymorphism of human peptidase C (PEPC)

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INTRODUCTION

Peptidase C (PEPC) was recently demonstrated to be polymorphic in man (Kömpf et al. 1989).

In this work we present data on the formal and population genetics of PEPC from SW Germany and NW Portugal and we discuss the use of this marker in paternity expertises.

MATERIAL AND METHODS

Blood samples were collected by venipuncture. Phenotyping was performed either in leucocytes or in red cell lysates, used fresh or after storage at -20°C up to 7 years. Sample treatment, electrophoretic separation and staining were performed as described previously (Kömpf et al. 1989).

RESULTS

The common phenotypes (1, 6-1 and 6) of PEPC are shown in Fig.1. In Fig.2 the rare phenotypes 2-1 and 6-2 are depicted.

Population genetics data from SW Germany and NW Portugal are presented in Table 1.

The results of the segregation analysis in nuclear families from SW Germany and NW Portugal are shown in Tables 2 and 3.

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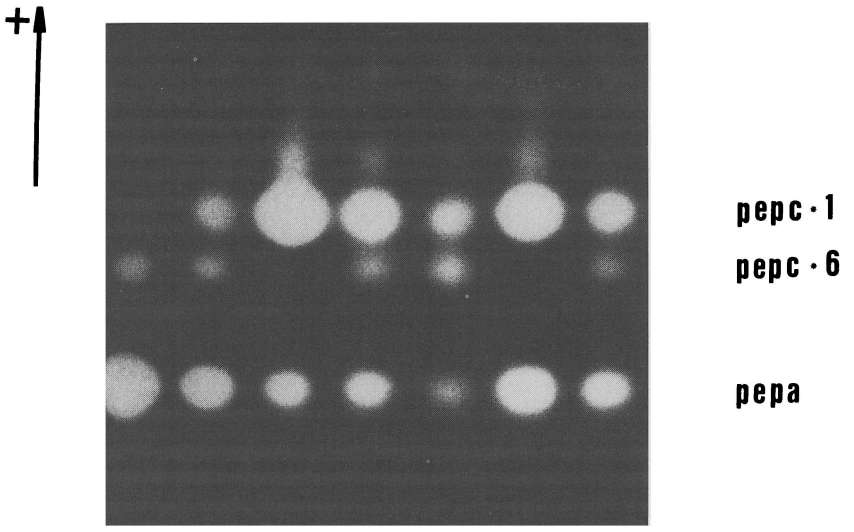

Fig.1. Common electrophoretic phenotypes of PEPC

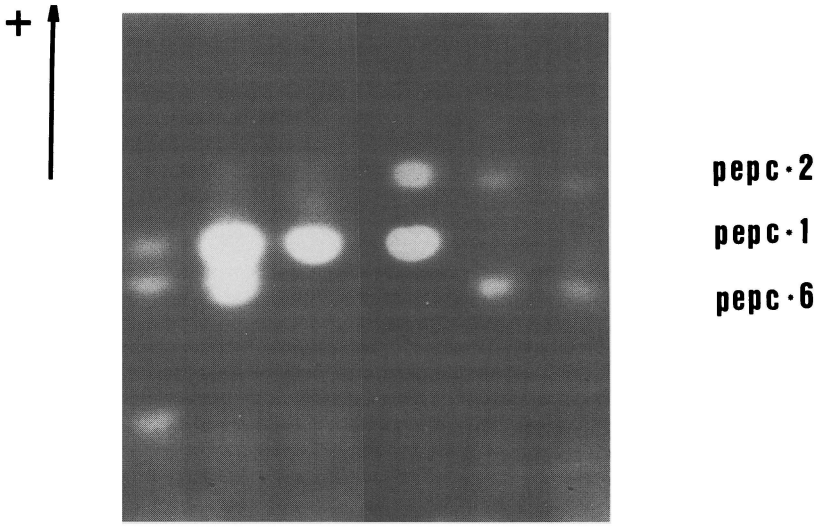

Fig.2. Rare electrophoretic phenotypes of PEPC

DISCUSSION

Electrophoretic patterns detected are consistent with a monomeric structure for PEPC (Figs 1 and 2). Apart from the common gene products PEPC*1 and PEPC*6, rare variants were found (Fig.2).

Table 1. PEPC phenotype distribution in SW Germany and NW Portugal

Population	N	phenotype 1	6-1	6	2-1	others	X^2
SW Germany	442	235 (235.5)	175 (174.2)	32 (32.3)	0	0	0.01
PEPC*1=.730 ; PEPC*6=.270							
NW Portugal	320	212 (206.3)	89 (100.2)	18 (12.2)	1 (0.3)	0 (0.0)	3.91
PEPC*1=.803 ; PEPC*6=.195 ; PEPC*2=.002							

Table 2. PEPC segregation analysis in family material from SW Germany

Mating type	nr. families	offspring 1	6-1	6	total	X^2
1 x 1	66	196			196	
1 x 6-1	92	130 (130.00)	130 (130.00)		260	0.0
1 x 6	11		29		29	
6-1x 6-1	35	35 (25.75)	40 (51.50)	28 (25.75)	103	6.0
6-1x 6	13		19 (19.50)	20 (19.50)	39	0.1
6 x 6	4			12	12	
total	221	361	218	60	639	

Segregation analysis (Tables 2 and 3) supports an autosomal genetic model with codominant alleles. However, in Portuguese material, evidence for a silent gene (2 families, PEPC*0=0.006) was found.

Table 3. PEPC segregation analysis in family material from NW Portugal

Mating type	nr. families	offspring 1	6-1	6	1-V	total
1 x 1	40	127				127
1 x 6-1	33	49	67	5		121
1 x 6	10	2	30			32
6-1x 6-1	7	6	14	6		26
1 x 1-V	1	2			1	3
total	91	186	111	11	1	309

Concerning population genetics, the observed phenotype distribution agrees well with Hardy-Weinberg expectations based upon a codominant model in the German sample. The fit is poor in the Portuguese material, with a deficiency of heterozygotes, probably due to the relatively common silent gene detected in family analysis.

Therefore it seems that PEPC can be reliably used in paternity expertises and, given the gene frequencies found in both populations, it proves to be a rather informative marker.

REFERENCE

Kömpf J, Prata MJ, Amorim A (1989) Genetic polymorphism of human peptidase C, PEPC (E.C. 3.4.1.1): formal genetic and population data. Hum Genet 83: 197-198