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Distribution of Plasma Genetic Markers in Central Portugal

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Resumo. Terceiro componente do complemento (C3), amilase pancreática (AMY2), haptoglobina (HP), componente específico de grupo (GC), transferrina (TF) e albumina (ALB) foram estudados em indivíduos não aparentados nascidos na região centro de Portugal. Os resultados obtidos foram: AMY2*1=0.960, AMY2*2=0.038, AMY2*3=0.0005, AMY2*4=0.0005 (N=1107); C3*F=0.202, C3*S=0.791, C3*Fv=0.001, C3*Sv=0.006 (N=564); GC*1=0.73, GC*2=0.27 (N=179); HP*1=0.39, HP*2=0.61 (N=384); TF*C=0.996, TF*B=0.003, TF*D=0.001 (N=974); ALB*A=0.9988, ALB*V_1=0.0006, ALB*V_2=0.0006 (N=853).

Palavras-chave: Genética populacional; Centro de Portugal; ALB, AMY2, C3, GC, HP, TF.

Abstract. Third component of complement (C3), pancreatic amylase (AMY2), haptoglobin (HP), group-specific component (GC), transferrin (TF) and albumin (ALB) were studied in unrelated individuals from central Portugal. The gene frequencies found were: AMY2*1=0.960, AMY2*2=0.038, AMY2*3=0.0005, AMY2*4=0.0005 (N=1107); C3*F=0.202, C3*S=0.791, C3*Fv=0.001, C3*Sv=0.006 (N=564); GC*1=0.73, GC*2=0.27 (N=179); HP*1=0.39, HP*2=0.61 (N=384); TF*C=0.996, TF*B=0.003, TF*D=0.001 (N=974); ALB*A=0.9988, ALB*V_1=0.0006, ALB*V_2=0.0006 (N=853).

Key Words: Population genetics; Central Portugal; ALB, AMY2, C3, GC, HP, TF.

Introduction

Serum protein polymorphisms demonstrated by electrophoretic techniques, are a valuable tool for population and paternity studies. However, very little has been reported concerning these genetic markers in Portuguese populations (Amorim, 1983; Vide, 1988; Torrinha, 1967; Weissman and Reuter, 1982).

In this paper, we present the results of a population study in central Portugal. The genetic markers analyzed were: third component of complement (C3), pancreatic amylase (AMY2), haptoglobin (HP) and group-specific component (GC). We also report the results on transferrin (TF) and albumin (ALB) rare variants.

Material and Methods

Blood samples from unrelated individuals living in central Portugal (districts of Aveiro, Castelo Branco, Coimbra, Guarda, Leiria, Santarém and Viseu) were obtained by venipuncture in EDTA tubes. The plasma was separated and stored at -20ºC until analysis.

The C3, TF and ALB phenotypes were determined by high voltage agarose gel electrophoresis, according the method described by Teisberg (1970). After running, the gels were stained with Comassie Brillant Blue. The albumin variants were then reanalyzed by IEF-PAGE according to Rocha *et al.* (1991).

GC phenotyping was performed by vertical polyacrylamide gel electrophoresis, using the method of Welch *et al.* (1979).

For AMY2 typing, gel electrophoresis was performed according to Kömpf *et al.* (1979) with some modifications (Amorim, 1983). After run, the gels were incubated with an agar-gel containing starch (1%), NaCl (0.5%) and Ca-lactate (0.06%) and stored at 37°C for about 2 hours. The gels were then stained in Lugol's solution.

Common HP phenotypes were detected by means of polyacrylamide gel electrophoresis, according to WELCH *et al.* (1979) and also in starch gel as described by Poulik (1957). For staining procedure, hemoglobin was added to serum samples. The HP bands appeared by means of hemoglobin peroxidase activity using 8.4mM phenylenediamine in 1M acetate buffer, pH 4.6, with some drops of commercial H_2O_2 solution.

Results and Discussion

The distribution of phenotypes and gene frequencies obtained are presented in Table 1. There was a good agreement between observed and expected values assuming Hardy-Weinberg equilibrium.

In previous investigations of portuguese populations we can found similar gene frequencies for AMY2, HP, GC, and TF systems (Amorim, 1983; Torrinha,

Table 1. Phenotype distributions and gene frequencies in Central Portugal.

Phenotypes		Observed	Expected	Gene frequencies
C3				
	F	19	23.04	C3*F 0.202±0.012
	FS	188	180.30	C3*S 0.791±0.012
	S	349	352.69	C3*Fv 0.001±0.001
	FvS	(*)1	0.79	C3*Sv 0.006±0.002
	FSv	(*)2	1.41	
	SSv	5	5.54	
	other	(*)—	0.23	
Total		564	564.00	χ^2=1.26 1df 0.3<p<0.5
AMY2				
	1	1024	1020.75	AMY2*1 0.9602±0.004
	2—1	76	82.58	AMY2*2 0.0388±0.004
	2	5	1.67	AMY2*3 0.0005±0.0004
	3—1	1	0.96	AMY2*4 0.0005±0.0004
	4—1	1	0.96	
	other	—	0.08	
Total		1107	1107.00	
HP				
	1	63	59.77	HP*1 0.39±0.01
	2—1	177	183.46	HP*2 0.61±0.01
	2	144	140.77	
Total		384	384.00	χ^2=0.48 1df 0.3<p<0.5
GC				
	1	94	95.87	GC*1 0.73±0.02
	2—1	74	70.26	GC*2 0.27±0.02
	2	11	12.87	
Total		179	179.00	χ^2=0.51 1df 0.3<p<0.5
TF				
	C	966	966.22	TF*C 0.996±0.001
	BC	6	5.82	TF*B 0.003±0.001
	CD	2	1.94	TF*D 0.001±0.001
	other	—	0.02	
Total		974	974.00	

(*) these classes were pooled for χ^2 calculation

1967; Vide, 1988; Weissman and Reuter, 1982). The C3 allele frequencies were foundto be quite similar (C3*F=0.20) to those obtained in Faro and Lisboa by Weissman and Reuter (1982) but different from those reported for Porto district by Amorim (1983) and central Portugal by Vide (1988): C3*F=0.15 and C3*F=0.17, respectively.

When comparing our results with those from Spanish neighboring areas, similar frequencies are found for all markers (Caeiro, 1987; Carracedo *et al.*, 1987; Carracedo and Concheiro, 1983; Goedde *et al.*, 1973; Moral *et al.*, 1988).

Two rare variants, classified as Sv and Fv, are found in C3 system (Fig.1). The frequency of Sv allele is in the bordline of polymorphic proportions (0.006), confirming the data obtained by Amorim (1983) in Porto district.

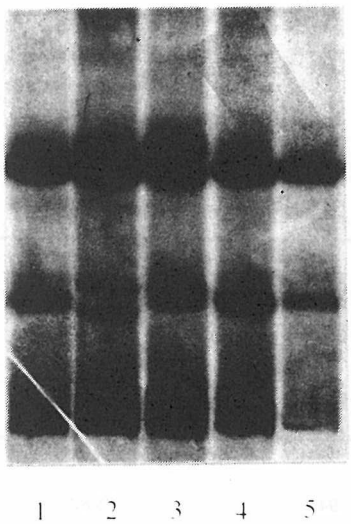

Fig. 1. *Common phenotypes and a rare variant of third component of complement (C3). Lanes 1,4,5=(S-S); 2=(F-Sv); 3=(F-S).*

In AMY2 system we also found the rare alleles AMY2*3 and AMY2*4 (Fig. 2) previously described by Kömpf *et al.* (1979).

The frequencies of TF rare alleles (Fig. 3), TF*B=0.003 and TF*D=0.001, are within the European ranges according to Walter (1980).

The non-identity of the two albumin variants detected (Fig. 3), was confirmed by IEF-PAGE (Rocha *et al.*, 1991) demonstrating that they correspond to different gene products.

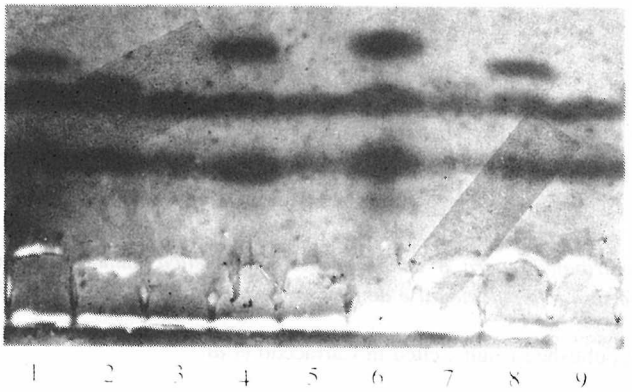

Fig. 2. Common phenotypes and rare variants of pancreatic amylase (AMY2). Lanes 1,8=(1-3); 2=(1-2); 3,5,7,9=(1-1); 4,6=(1-4).

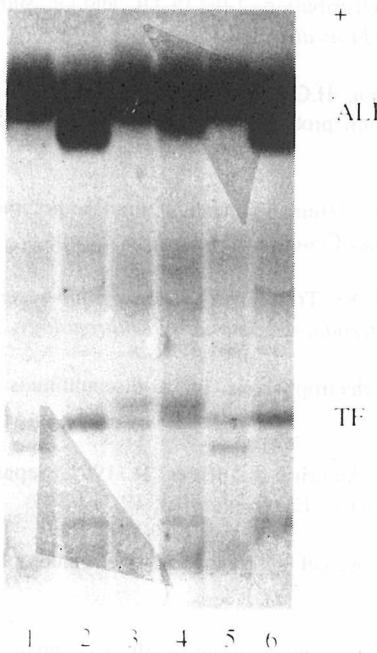

Fig. 3. Common phenotypes and rare variants of transferrin (TF) and albumin (ALB) after high voltage agarose gel electrophoresis. TF: Lanes 1,5=(CD); 2,6=(CC); 3,4=(BC). ALB: Lanes 1,3,5=(AA); 2,6=(AV_2); 4=(AV_2).

In conclusion, we have demonstrated (a) the usefulness of these markers for anthropological and applied population genetics (namely paternity expertises) and (b) the genetic homogeneity of the population under study when compared with Northern Portugal. The only relevant difference found (in C3) is under study in a larger sample, in order to confirm if there is, indeed, a North-South cline in this system.

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