

GC, TF AND PI POLYMORPHISMS IN MADEIRA (PORTUGAL) PI*S ALLELE AND PATERNITY TESTING

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INTRODUCTION

Madeira is an island in the Atlantic Ocean located about 500 miles off the southwest coast of Portugal, where the Portuguese, since the 15th century, contributed mainly to the present-day population.

The primary purpose of the present paper was to study the distribution of GC, TF and PI phenotypes in Madeira to obtain a estimate of allele frequencies for paternity testing purposes, with special reference to the PI*S allele frequency due to its great interest for population genetic studies.

MATERIAL AND METHODS

Serum samples from a total of 151 unrelated individuals from Madeira were collected from paternity tests.

GC, TF and PI phenotyping was carried out by isoelectric focusing with carrier ampholytes on flat-bed polycrylamide gels. GC system was studied according to the method described by Constans and Viau (1) with slight modifications. GC bands were made visible after precipitation with sulfosalicylic acid. PI system was typed according to Hjalmarsson (2). For TF phenotyping, serum samples were analysed as indicated by Budowle (3). Staining of TF and PI gels was carried out with Coomassie Blue R 250

RESULTS AND DISCUSSION

Table 1 shows the distribution of GC, TF and PI phenotypes and their allele frequencies for the population from Madeira. A good agreement was found between observed and expected number assuming a Hardy-Weinberg equilibrium.

The theoretical a priori chance exclusion values for these systems are : GC= 0.2984, TF= 0.1872 and PI= 0.3807, which indicate that these systems are useful genetic markers for paternity investigation in the population from Madeira.

The allele frequencies for GC and TF systems are not significantly different from our results (not published) of previous studies of the Continental Portuguese population, mainly from the South.

Table 1. Distribution of GC, TF and PI phenotypes and allele frequencies in Madeira

Phenotypes		Observed Number	Expected Number	Allele Frequencies	
GC System	1S	45	47.85		
	2-1S	57	51.79	GC*1S= 0.5629	
	1S-1F	23	22.52	GC*1F= 0.1325	
	2	12	14.01	GC*2 = 0.3046	
	2-1F	11	12.19		
	1F	3	2.65		
Total		151	151.00		
$\chi^2 = 1.15$; df=3; $0.70 < p < 0.80$					
TF System	C1	89	88.35		
	C1C2	40	42.07	TF*C1= 0.7649	
	C1C3	12	11.47	TF*C2= 0.1821	
	C1B	1	0.76	TF*C3= 0.0497	
	C1C2	6	5.01	TF*B = 0.0033	
	C2C3	3	2.73		
	C2B	0	0.18		
	C3	0	0.37		
	C3B	0	0.05		
	B	0	0.00		
	Total		151	151.00	
	$\chi^2 = 1.03$; df= 6; $0.98 < p < 0.99$				
	PI System	M1	50	47.85	
M1M2		22	25.33		
M1M3		18	17.45	PI*M1=0.5629	
M1S		29	30.96	PI*M2=0.1490	
M1Z		1	0.56	PI*M3=0.1027	
M2		5	3.35	PI*S = 0.1821	
M2M3		6	4.62	PI*Z = 0.0033	
M2S		7	8.20		
M2Z		0	1.59		
M3S		3	5.65		
M3Z		0	0.10		
S		8	5.01		
SZ		0	0.18		
Z		0	0.00		
Total		151	151.00		
$\chi^2 = 5.97$; df=10; $0.80 < p < 0.90$					

In the PI system, we have observed a higher allele frequency for PI*S allele ($f= 0.1821$) in Madeira when compared to the Continental Portuguese population ($f= 0.1141$) and even when compared to Southern European populations from Spain and France ($f= 0.1044$ to 0.1490 - table 2). Consequently, PI*M1 allele frequency is lower in Madeira.

Table 2 PI*S allele frequencies in several Southern European populations

Population	Number of Subjects	Allele Frequency PI*S	References
Portugal/Madeira	151	0.1821	This study
Mainly South	1850	0.1140	Not published
Spain/Galicia	450	0.1490	Carracedo and Concheiro (4)
Barcelona	938	0.1044	Gènè et al (5)
Basques	323	0.1006	Manzano et al (6)
France /Southwest	1603	0.1095	Sesboué et Martin (7)

The highest frequency for PI*S allele have been found in the Galicia population (4) . Our study supports the European cline described for the PI*S allele from Northern to Southern European populations (8), reaching a maximum in Madeira.

REFERENCES

1. Constans J, Viau M: Group-specific component: Evidences for two subtypes of the GC gene. *Science* 198:1070-107 (1977)
2. Hjalmarsson K : Distribution of alpha-1-antitrypsin phenotypes in Sweden. *Hum Hered* 38: 27-30 (1988).
3. Budowle B: Improved separation of the common transferrin variants in gels containing pH5-7 ampholines and HEPES. *Electrophoresis* 8: 210-212 (1987).
4. Carracedo A, Concheiro L: Distribution of the PI, TF and GC Subtypes in Galicia (North West Spain). *Z Rechts med.* 90: 153-158 (1983).
5. Genè M, Huguët E, Carracedo A, Ercilla G, Corbella J: Frequency and distribution of PI, GC, TF, and PLG subtypes by isoelectric focusing in Barcelona. *Advances in Forensic Haemogenetics*, Vol. 1, pp 235-239 (Springer-Verlag, Berlin, Heidelberg 1986).
6. Sesboué R, Martin J P : Alpha-1-antitrypsin (PI) polymorphism in France, with special regard to the PI*Z allele. *Hum Hered.* 41: 340-346 (1991).
7. Fagerhol M K , Cox D W: The Pi polymorphism genetic, biochemical and clinical aspects of human alpha-1-antitrypsin. *Adv. Hum. Genet.* 11: 1-62 (1981).