

ISOELECTRIC FOCUSING OF INTER-ALPHA-TRYPSIN INHIBITOR (ITI)

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INTRODUCTION

The genetic polymorphism of human plasma inter-alpha-trypsin inhibitor (ITI), a serine protease inhibitor, was first described by Vogt and Cleve (1990). Both precise subunit composition and chromosomal assignment of the corresponding locus remain under investigation (Schreitmüller et al., 1987, Luckenbach et al., 1991). In this work we present formal and population genetics results obtained in families from SW Germany and NW Portugal.

MATERIAL AND METHODS

Blood samples were obtained by venipuncture and EDTA plasmas stored at -20°C until use. Sample treatment and phenotyping were performed as previously described (Luckenbach et al, 1991).

RESULTS AND DISCUSSION

Phenotypes of ITI are shown in Fig.1; nomenclature was made according to Vogt and Cleve (1990). Banding patterns (a main band for homozygotes and two for heterozygotes) in non-denaturing conditions are consistent with a monomeric protein. However, given the presence of additional weaker bands and the possibility of loose association between monomeres, we can not rule out more complex quaternary structures, as suggested by Schreitmüller et al.(1987)

In Tables 1 and 2 we show the mating type distribution found in SW Germany and NW Portugal. Both distributions agree well with Hardy-Weinberg expectations. They clearly support both qualitatively and quantitatively the formal hypothesis for ITI: an autosomal locus with two common codominant alleles (ITI*1, ITI*2) and a rare one, also codominant (ITI*3). No evidence of silent genes was found.

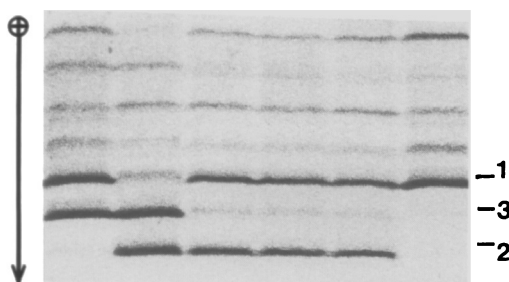


Fig.1. ITI phenotypes.

From left to right:

3-1; 3-2; 2-1; 2-1; 2-1
and 1

TABLE 1. ITI segregation analysis. Families from SW Germany

mating type	no families	offspring phenotypes					Allele frequencies	
		1	2-1	3-1	2	3-2	ITI*1	ITI*2
1 x 1	39 32.38	103					0.607	
1 x 2-1	75 82.85	91	101				0.388	0.005
1 x 3-1	2 1.12	3		3				
1 x 2	32 26.50		105					
2-1x 2-1	47 52.99	43	72		28			
2-1x 2	34 33.90		51		50			
2-1x 3-2	1 0.91		0	1	0	2		
2 x 2	7 5.42		0.75	0.75	0.75	0.75		
2 x 3-2	2 0.29				4	3		
Others	0 2.64				3.5	3.5		
Total	239	240	329	4	99	5		

TABLE 2. ITI segregation analysis. Families from NW Portugal

mating type	no families	offspring phenotypes			Allele frequencies	
		1	2-1	2	ITI*1	ITI*2
1 x 1	2 2.69	8			0.552	0.448
1 x 2-1	10 8.73	12	16			
1 x 2	3 3.55		9			
2-1x 2-1	8 7.10	9	7	2		
2-1x 2	4 5.77	4.5	9.0	4.5		
2 x 2	2 1.17		5.0	5.0		
Total	29	29	39	10		

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