

Formal genetics studies on ALAD, AMY2, ESD, GLO1, HP, ORM and PGP polymorphisms

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The genetic polymorphisms of ALAD (aminolevulinatase, E.C.4.2.1.24), AMY2 (pancreatic amylase, E.C.3.2.1.1), ESD (esterase D, E.C.3.1.1.1), GLO1 (glyoxalase I, E.C.4.4.1.5), HP (haptoglobin), ORM (orosomuroid) and PGP (phosphoglycollate phosphatase, E.C.3.1.3.18) are currently used in paternity expertises.

The purpose of this work is to perform a revision of the formal genetics of these systems, using family material from SW Germany and NW Portugal.

MATERIAL AND METHODS

Phenotyping methods are described in previous publications of the research groups involved.

RESULTS AND DISCUSSION

In Table 1 we present sample data and calculated gene frequencies for each marker in SW Germany and NW Portugal.

ALAD

No mother/child exclusions were found.

The observed segregation results agree well with expected ones.

AMY2

Two mother/child exclusions were found, one in each sample (estimated AMY2*O frequencies: 0.001 in SW Germany and 0.002 in NW Portugal).

On the other hand, the segregation in the offspring of mating type 1x2-1 is clearly distorted (Table 2).

ESD

No mother/child exclusions were found.

The observed segregation results agree well with expected ones, if the sex of children is not taken into account. However, a significant sex/phenotype association was found in both samples when analysing mating types 1x2-1 and its reciprocal.

GL01

Two mother/child exclusions were found, one in each sample (estimated GL01*O frequencies: 0.001 in SW Germany and 0.002 in NW Portugal).

Concerning the segregation analysis, in the Portuguese material the offspring of 2-1x2-1 mating type show a significant deviation from expected values ($X^2=6.55$, 2 d.f.); in the German sample a general trend to homozygote excess is also observed (Table 3). On the other hand, in the Portuguese sample the offspring of mating type: father 2-1 x mother 1 show a significant sex/phenotype association.

HP

A mother/child exclusion was found in the Portuguese sample (estimated HP*O frequency: 0.002).

Apart from that, the observed segregation results agree well with expected ones.

ORM

No mother/child exclusions were found.

The observed segregation results agree well with expected ones, but in the Portuguese sample a significant deviation was found in mating type 1x2-1 (16 vs. 6 children respectively 1 and 2-1). The small sample size prevents us from giving relevance to this finding.

PGP

No mother/child exclusions were found.

Concerning segregation analysis, a significant distortion was found in both samples for mating type: father 2-1 x mother 1, but not in its reciprocal (Table 4).

CONCLUSIONS

The results here reported confirm the need for regular formal genetics researches on all genetic systems used in paternity expertises, including those for which the formal genetic model is considered to be established.

Furthermore, the fact that in some of the markers here studied the detected segregation disturbances were present in independent samplings geographically rather distant, seems to substantiate the conclusion that at least for these markers the deviations are not due to mere chance and deserve a further investigation.

TABLE 1 Gene frequencies and sample sizes used for segregation analysis. G and P stand respectively for SW Germany and NW Portugal.

System	Sample	Nr.		Frequencies of the alleles (%)				
		fam.	child.	1	2	3	5	V
ALAD	G	222	629	92	8			
	P	133	263	89	11			
AMY2	G	258	784	97	3			
	P	108	230	94.2	5.6	0.2		
ESD	G	176	494	86.2	12.5		1.3	
	P	140	486	86.1	13.0		0.7	0.2
GLO1	G	210	697	43	57			
	P	109	242	44	56			
HP	G	91	268	40	60			
	P	110	247	39	61			
ORM	G	159	479	61	39			
	P	35	47	61	39			
PGP	G	184	486	87	10	3		
	P	113	242	92	5	3		

TABLE 2 AMY2 segregation analysis. Family material from SW Germany and NW Portugal.

mating type		Nr.		Phenotypes of the children		
male	x female	fam.	child.	1	2-1	X2
1	2-1	19	67	46	21	9.33
2-1	1	30	81	49	32	3.57
TOTAL		49	148	95	53	11.92

TABLE 3 GLO1 segregation analysis. Family material from SW Germany.

mating type male/female	Nr.		Phenotypes of the children			X ²
	fam.	child.	1	2-1	2	
1 x 2-1	29	82	37	45		.780
2-1 x 1	34	102	45	57		1.412
2-1 x 2-1	81	240	51	133	56	3.025
2-1 x 2	63	169		95	74	2.609
2 x 2-1	56	190		113	77	6.821

TABLE 4 PGP segregation analysis. Family material from SW Germany and NW Portugal.

mating type male x female	Nr.		Phenotypes of the children			X ²
	fam.	child.	1	2-1	2	
1 x 2-1	22	46	18		28	1.562
2-1 x 1	24	70	46		24	6.914
TOTAL	46	116	64		52	1.241