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Studies on the placental alkaline phosphatase and Phosphoglucomutase (PGM_1) polymorphisms in the province of Pavia (Italy)

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Genetica. — *Studies on the placental alkaline phosphatase and Phosphoglucomutase* (PGM_1) *polymorphisms in the province of Pavia (Italy)*. Nota di RICCIOTTI PALMARINO (*), RENATO SCACCHI (*), ROSA MARIA CORBO (*), EMILIO IMPARATO (**), PAOLO PESANDO (**) e PAOLA LUCARELLI (*), presentata (***) dal Socio G. MONTALENTI.

RIASSUNTO. — È stato determinato il fenotipo della fosfatasi alcalina della placenta e della fosfoglucomutasi in un gruppo di soggetti della provincia di Pavia. Le frequenze geniche relative alla fosfoglucomutasi, locus 1, non sono risultate differenti da quelle osservate nelle altre popolazioni italiane, mentre per la fosfatasi alcalina placentare sono state osservate alcune differenze. Sono stati inoltre individuati sei fenotipi rari per la fosfatasi alcalina placentare.

The human placenta has an elevated content of alkaline phosphatase which differs from that of other organs in various of its physico-chemical properties [1-2-14-15-22]; more over it is the only alkaline phospatase for which an electrophoretic polymorphism has been demonstrated [14-23-24]. By starch gel electrophoresis at two different pH (6.0 and 8.6) Robson and Harris were able to demonstrate 6 common electrophoretic patterns of the enzyme which account for 98% of the population; about 2% of the placentae revealed different patterns. Their frequencies are in agreement with the hypothesis that they are determined by three common codominant alleles at one autosomal locus. The enzyme is of foetal origin; it is present in the serum of almost all women by the 29th week of gestation and disappears by the 6th week after delivery. Wide interracial variation in the frequency of Pl alleles has been reported [3-4-5-7-8-9-10-18-19]. The biological significance of this polymorphism is not clear but there are now some observations which suggest that the enzyme plays an important role in the maternal-fetal interactions [6-12-13].

The phosphoglucomutase is an ubiquitous enzyme which reversibly catalyzes the transfer of phosphate from the first to the sixth position of glucose. By starch gel electrophoresis multiple isozymes of human phosphoglucomutase can be detected. They appear to be due to alleles at three distinct and not closely linked autosomal loci each of which determines a separate group of PGM isozymes (PGM_1, PGM_2 and PGM_3) [16-17-26]. The population frequencies and the genetics of the PGM isozymes attributed to the PGM_1 locus have been usually worked out using red cells. Since we had demonstra-

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ted [20] that the placental PGM_1 phenotypes are controlled exclusively by the fetal genotype, we have used specimens of human placenta for the determination of both Pl and PGM_1 phenotypes.

Materials and Methods

Placentae were obtained from consecutive deliveries at the Obstetric Clinic of the University of Pavia and kept frozen until they were analyzed. The mother and father of the infants were interviewed in regard to the origin of their ancestors and only those subjects whose four grandparents were original of the province of Pavia were examined.

Alkaline phosphatase: the placental extracts were prepared as described by Boyer [15]; starch gel electrophoresis at pH 6.0 and 8.6 were performed according to Robson and Harris [22] and enzymatic activity was developed by the method of Boyer [14].

Phosphoglucomutase: tissue extracts were prepared by homogenizing a small volume of tissue with an equal volume of distilled water. The omogenates were centrifugated at about 10,000 r.p.m. and the clear supernatant was used for electrophoresis. Starch gel electrophoresis was performed according to Spencer *et al.* [26]. After electrophoresis PGM components were visualized as described by Spencer *et al.* [26].

Results and discussion

Tables I and II show the phenotypes and gene frequencies of Pl and PGM_1 polymorphisms. There is a close correspondence with the frequencies expected by the Hardy-Weinberg equilibrium.

Table I.

Observed and expected frequencies of the six common human placental alkaline phosphatase phenotypes in the population of Pavia (Rare phenotypes are not included).

Phenotypes	S	F	I	SF	SI	FI	Totals
Observed . . .	70	10	4	65	21	10	180
Expected . . .	70.97	12.53	2.09	59.65	24.41	10.26	179.91
χ^2	0.013	0.511	1.745	0.480	0.476	0.006	3.231

Gene frequencies: Pl^{s1} 0.628; Pl^{f1} 0.264; Pl^{i1} 0.108.

TABLE II.

Observed and expected frequencies of phosphoglucomutase (locus 1) phenotypes in the population of Pavia.

Phenotypes	1–1	2–1	2–2	Totals
Observed	98	73	19	190
Expected	95.23	78.55	16.19	189.97
χ^2	0.080	0.392	0.488	0.960

Gene frequencies: PGM_1^1 0.708; PGM_1^2 0.292.

TABLE III.

Human placental alkaline phosphatase gene frequencies in different italian populations.

Italian populations	Totals	Gene frequencies			References
		Pl^{s_1}	Pl^{f_1}	Pl^{i_1}	
Pavia	190	62.8	26.4	10.8	This paper
Rome	546	66.1	25.6	7.5	[11]
L'Aquila	378	57.7	34.1	7.1	[11]

TABLE IV.

Phosphoglucomutase (locus 1) gene frequencies in different italian populations.

Italian populations	Totals	Gene frequencies PGM_1^1	References
Pavia	190	70.8	This paper
Rome	389	71.2	[21]
Sardinia	1641	73.0	[27]
L'Aquila	385	70.3	[25]

The PGM_1 gene frequencies found in the present sample are similar to those found in the population of Rome, L'Aquila and Sardinia.

As far as the human placental alkaline phosphatase, while the frequencies of the 3 common alleles in the population of Pavia are quite similar to those found in Rome and in some North-European populations, they are signi-

ficantly different ($\chi^2_{2\,d.f.} = 7.281$: $P = \sim 0.025$) from those observed in the population of L'Aquila.

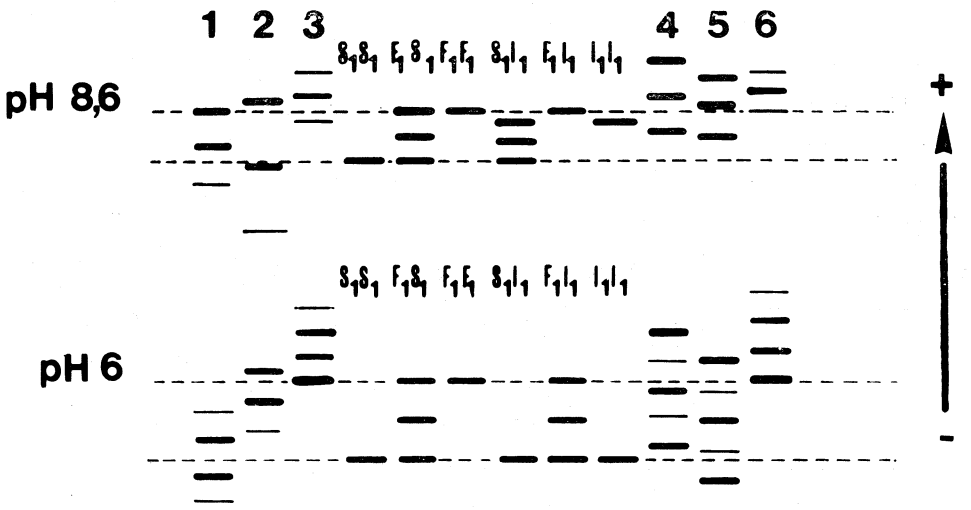

Fig. 1 – Diagram of starch-gel electrophoretic separation of different placental alkaline phosphate phenotypes.

Six rare phenotypes of human placental alkaline phosphatase were detected (3.3%). All the phenotypes observed are reported in fig. 1, at both pH 8.6 and 6.0.

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