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ISOELECTRIC FOCUSING (IEF) OF HUMAN RED CELL PHOSPHOGLUCOMUTASE (PGM1). THE DISTRIBUTION OF PHENOTYPES IN NORTH POLISH POPULATION[1]

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Summary. The human PGM erythrocyte polymorphism was first described by Spencer et al. (1964). Using starch gel electrophoresis they demonstrated two codominant alleles. Recent studies using technique of IEF on different gels have shown that PGM1 is coded by four autosomal codominant alleles: PGM1*1A, PGM1*1B, PGM1*2A, and PGM1*2B (Bark et al. 1976, Kühnl et al. 1977, Sutton, Burges 1978). The evidence of four alleles and 10 common phenotypes was also demonstrated using conventional electrophoresis in acidic pH (Bissbortt et al. 1978, Bär, Dissing 1983, Wolson, Stuver 1985). This paper presents data on the distribution of PGM1 subtypes in North Polish population (Gdańsk).

MATERIAL AND METHODS

Blood samples were collected from 517 unrelated persons. Hemolysates were prepared from washed and packed red cells by dilution with redistilled water. After freeze-thaw cycle hemolysates were stored at −20°C until use. Polyacrylamide gels (5% T, 3% C, 200 × 12 × 0.2 mm) were cast onto silanised glass plates using "flap technique" of Radola (1980). Gels containing 3% mixture of carrier ampholines (two parts of Ampholine pH 5-7 and one part of Servalyt pH 5-7) and 1.2% EPPS as a separator were polymerized using ammonium persulfate. The anolyte and catolyte were 0.9M H3PO4 and 1M NaOH, respectively. The electrode distance was 9 cm. The IEF was conducted on the Ultrophor (LKB, Bromma, Sweden) at 6°C. The gels were prefocused at a constant power of 2W for 25 min. Samples were applied 1 cm from the anodal electrode strip. After 10 min. tubs were removed and gels were run at a constant power of 6W for

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60 min. at maximum 2500V and unlimited current. The constant power mode during all run was applied as recommended by Budowle (1984). Visualization of the isozyme bands patterns was carried out according to the method of Divall and Ismail (1983).

RESULTS AND DISCUSSION

Fig. 1 shows the IEF pattern of six out of the ten common PGM1 subtypes detected in the studied population. Owing to the EPPS application and carrier ampholines mixture the 2A, 2B, 1A and 1B bands were well saturated with a 2A to 1B distance of 32 mm. Moreover, the application, of the ultra-thin layer gel combines high resolution, speed and economy with simplicity of operation.

Table 1. Frequencies of PGM1 phenotypes and alleles in North Polish population sample of 517 unrelated adults

Phenotype	Observed		Expected		x^2
	N	%	N	%	
1A	192	37.14	194.37	37.59	0.0289
1B	9	1.74	8.42	1.63	0.0399
1A1B	87	16.83	80.94	15.65	0.4537
2A	25	4.83	20.53	3.97	0.9732
2B	1*	0.19	1.86	0.36	1.4547
2A2B	13	2.51	12.35	2.39	0.0342
2A1A	121	23.42	126.31	24.43	0.2232
2A1B	22	4.25	26.30	5.08	0.7030
2B1A	42	8.12	38.01	7.35	0.4188
2B1B	5*	0.97	7.91	1.55	
Total	517	100.00	517.00	100.00	4.3296

* pooled in x^2 – test; d.f.=8; $0.80 < P > 0.90$

Allele frequencies: PGM1*1A=0.6131; PGM1*1B=0.1276; PGM1*2A=0.1992; PGM1*2B=0.0600

Table 1 presents the distribution of the observed and expected data of PGM1 subtypes in the examined population. The observed numbers of phenotypes were in good agreement with the numbers expected under the equilibrium condition ($x^2=4.3296$, d.f.=8, $0.8<P>0.9$). The estimated gene frequencies were PGM1*1A=0.6131, PGM1*2A=0.1992 and PGM1*2B=0.0600.

Table 2 presents reported gene frequencies of the PGM1 subtypes in different European populations. Data from this study (Table 1) and reported by others for the different European populations and South district of Poland (Kraków) show a similar frequency of the four common alleles.

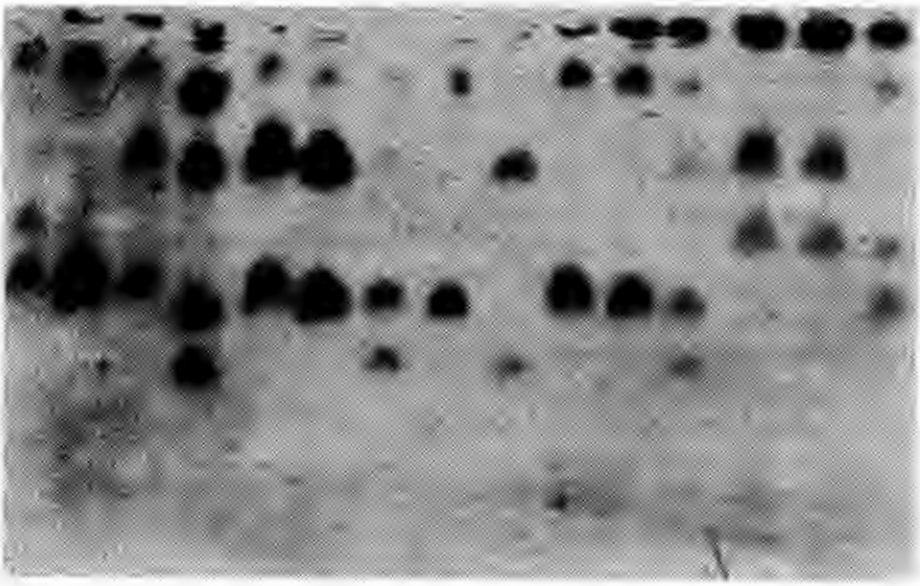

Fig. 1. A 0.2 mm thick polyacrylamide gel with 1.2 % EPPS and mixture of Ampholine pH 5-7 and Servalyt pH 5-7 (2:1), displaying PGM1 subtypes of fresh hemolysates. Phenotypes from left to right: 2B1A, 1A, 2A1A, 1A1B, 2A1A, 2A1A, 1A1B, 1A, 2A1B, 1A, 1A, 1A1B, 2A2B, 2A2B and 2B1A. Anode on the top

Table 2. Comparison of the reported gene frequencies of the PGM1 subtypes in the various european populations

Population	N	Gene frequencies				References
		PGM1*1A	PGM1*1B	PGM1*2A	PGM1*2B	
England	123	0.6341	0.1138	0.1829	0.0691	Bark et al. (1976)
	329	0.6367	0.1094	0.1778	0.0759	Welch et al. (1979)
Czecho-slovakia	495	0.639	0.118	0.180	0.063	Ranzani et al. (1985)
France	220	0.6636	0.1046	0.1750	0.0568	Vergnes, Sevin (1981)
Germany	291	0.6186	0.1426	0.1718	0.0670	Kühnl et al. (1977)
	620	0.6266	0.1895	0.1363	0.0476	Martin (1979)
	470	0.6212	0.1224	0.2043	0.0521	Schwarzfischer, Weideinger (1980)
	1678	0.6305	0.1320	0.1844	0.0530	Kühnl, Spielman (1978)
Italy	348	0.600	0.110	0.230	0.060	Scacchi et al. (1983)
	519	0.6012	0.1059	0.2495	0.0434	Bargagna, Abbagnale (1982)
	589	0.6180	0.1163	0.2122	0.0535	Cortivo et al. (1984)
	2975	0.594	0.118	0.231	0.057	Ranzani et al. (1985)
Spain	1086	0.621	0.114	0.211	0.054	Carracedo, Concheiro (1982)
Sweden	2000	0.61	0.16	0.16	0.07	Svensson, Wetterling (1979)
Switzer-land	501	0.6278	0.1297	0.1936	0.0489	Scherz et al. (1981)
	521	0.6324	0.1382	0.1939	0.0355	Bär, Dissing (1983)
Poland						
Kraków	460	0.6210	0.1185	0.1880	0.0533	Turowska, Nowicka (1987)
Gdańsk	517	0.6131	0.1276	0.1990	0.0600	This study

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IZOELEKTROOGNISKOWANIE FOSFOGLUKOMUTAZY (PGM1) ERYTROCYTÓW LUDZKICH. CZĘSTOŚCI FENOTYPÓW W POPULACJI POLSKI PÓŁNOCNEJ

Streszczenie

Izoelektroogniskowanie PGM1 przeprowadzono na ultracienkim żelu poliakrylamidowym w gradiencie pH 5-7 zawierającym EPPS jako separator. Dla 517 próbek krwi pobranych od niespokrewnionych osób uzyskano następujące częstości genowe: PGM1*1A=0,6131, PGM1*1B=0,1276, PGM1*2A=0,1992, PGM1*2B=0,0600. Porównanie uzyskanych wyników z częstościami genowymi w innych populacjach europejskich wykazało podobną częstość czterech powszechnie spotykanych alleli PGM1.

ИЗОЭЛЕКТРОФОКУСИРОВАНИЕ ФОСФОГЛЮКОМУТАЗЫ (PGM1) ЭРИТРОЦИТОВ ЧЕЛОВЕКА. ЧАСТОТЫ ФЕНОТИПОВ В ПОПУЛЯЦИИ СЕВЕРНОЙ ПОЛЬШИ

Резюме

Изоэлектрофокусирование PGM1 производилось на ультратонком полиакриламидном геле в градиенте pH 5-7, содержащим EPPS в качестве сепаратора. Для 517 проб крови, побранных у несвязанных родством особей, получено следующие генные частоты: PGM1*1A=0,6131, PGM1*1B=0,1276, PGM1*2A=0,1992, PGM1*2B=0,0600. Сравнение полученных результатов с генными частотами в других европейских популяциях показало, что частота четырёх широко выступающих аллелей PGM1 есть подобна.