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PGM_1^a Subtyping in Western Germany (Düsseldorf Region)

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Summary. Phosphoglucomutase (PGM_1^a) subtypes were determined by isoelectric focusing on samples of 496 unrelated individuals. Ten phenotypes were observed as gene products of four alleles at the PGM_1 locus, with the following gene frequencies: $PGM_1^{a1} = 0.631$, $PGM_1^{a2} = 0.194$, $PGM_1^{a3} = 0.126$, and $PGM_1^{a4} = 0.049$. A rare phenotype PGM_1 (8-a2) was observed.

Key words: Blood groups, phosphoglucomutase (PGM_1) – Phosphoglucomutase subtypes

Zusammenfassung. Die Phosphoglucomutase (PGM_1^a)-Untergruppen wurden durch isoelektrische Fokussierung aus Hämolysaten von 496 nicht verwandten Personen bestimmt. Zehn Phänotypen konnten beobachtet werden, die die Genprodukte der vier Allele des PGM_1-Genortes darstellen und folgende Genfrequenzen aufwiesen: $PGM_1^{a1} = 0{,}631$, $PGM_1^{a2} = 0{,}194$, $PGM_1^{a3} = 0{,}126$ und $PGM_1^{a4} = 0{,}049$. Ein seltener Phänotyp PGM_1 (8-a2) wurde beobachtet.

Schlüsselwörter: Blutgruppen, Phosphoglucomutase (PGM_1) – Phosphoglucomutase-Untergruppen

Introduction

The polymorphism of human phosphoglucomutase (E.C.: 2.7.5.1.) was first detected by Spencer et al. (1964).The ten variant gene products of the PGM_1 locus can be demonstrated either using the method based on isoelectric focusing on polyacrylamid gels (Bark et al. 1976; Kühnl et al. 1977; Kühnl and Spielmann 1978; Sutton and Burgess 1978; Welch et al. 1978) or by acid starch gel electrophoresis (Bissbort et al. 1978). We present here the gene frequencies of PGM_1^a in a population of Western Germany (Düsseldorf region). Furthermore, a rare phenotype could be detected.

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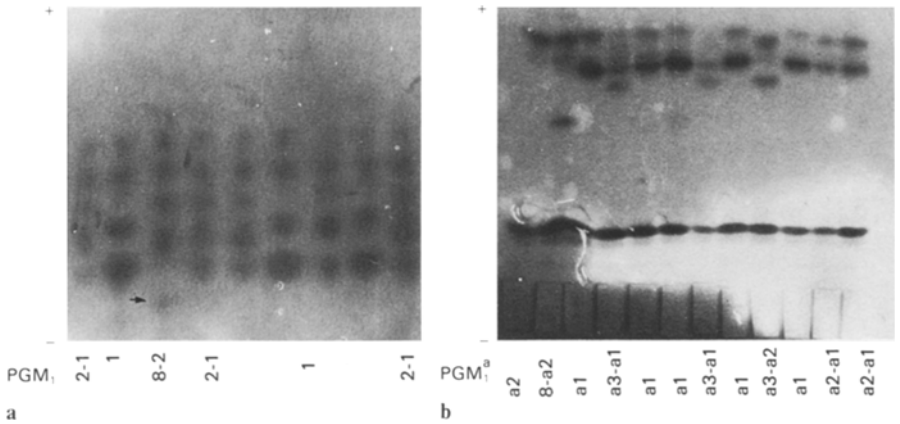

Fig. 1 a, b. PGM_1^a-subtyping

Materials and Methods

Red blood cells of 496 apparently healthy and unrelated individuals were treated with toluol and then sonified.

Isoelectric focusing (IEF) was performed with a LKB Multiphore electric focusing apparatus on PAG plates (LKB) of 2 mm thickness, pH range 3.5–9.5. The electrode paper strips were soaked with 1 *M* NaOH for the cathode and 1 *M* H_3PO_4 for the anode, respectively. After a prefocusing time of 1 h pieces of filter paper (Whatman No. 1, 5 × 10 mm) were placed on the gel 2 cm from the cathodal electrode strips, and 2 µl of water-diluted hemolysate (1:2, v/v) was added. Using an LKB 2103 power supply an electric current was applied for 3 h; the initial voltage was approximately 300 V (limited to 1,200 V) according to 70 mA. The power was stabilized at 22 W, and the plate was cooled to 3° C. The starch gel electrophoresis was performed according to Radam (1977). The enzyme pattern was made visible using the staining procedure described by Spencer et al. (1964).

Results and Discussion

In addition to some of the phosphoglucomutase (PGM_1) subtypes a rare PGM_1 (8-2a) is shown in Fig. 1 a after starch gel electrophoresis and in Fig. 1 b after isoelectric focusing. In the starch gel the isoenzyme activity of the PGM_1 8 allele is lower and can easily be ignored. By IEF the isoenzyme PGM_1 8 band is clearly detectable cathodically from the PGM_1 a3 band.

The distribution of the ten common PGM_1^a phenotypes in Western Germany is presented in Table 1 and the thus far reported gene frequencies of other German populations were compared with our results. The population analyzed here apparently underlies Hardy-Weinberg conditions. In comparison to the population studies in Hessen (Kühnl and Spielmann 1978), in the southwest of Germany (Bissbort et al. 1978) and in the northern part of Germany (Martin 1979; Simeoni and Grüner 1980) the gene frequencies are slightly but not statistically different.

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Table 1. Distribution of PGM$_1^a$-subtypes in Western Germany (Düsseldorf Region) and the comparison with the gene frequencies found in other parts of Germany

	PGM$_1$-Phenotypes									
	a1	a2-a1	a3-a1	a4-a1	a2	a3-a2	a4-a2	a3	a4-a3	a4
Observed	201	115	86	23	20	22	15	5	7	2
Expected	197.40	121.43	78.87	30.67	18.67	24.25	9.43	7.87	6.12	1.19
Total	$n=496$, $\chi^2=2.3556$, $0.70>P>0.50$, df 4									

		Gene frequencies					
	n	PGM$_1^{a1}$	PGM$_1^{a2}$	PGM$_1^{a3}$	PGM$_1^{a4}$	PGM$_1^{a1\backslash a3}$	PGM$_1^{a2\backslash a4}$
Kühnl and Spielmann (1978)	1.678	0.6305	0.1844	0.1320	0.0530	0.7625	0.2374
Bissbort et al. (1978)	329	0.650	0.175	0.123	0.052	0.773	0.227
Martin (1979)	620	0.6266	0.1895	0.1363	0.0476	0.7629	0.2371
Simeoni and Grüner (1980)	1.113	0.640	0.178	0.126	0.056	0.766	0.234
This paper	496	0.631	0.194	0.126	0.049	0.757	0.243

Phenotypes with n (expected) below 10 were combined for χ^2 calculations, the rare phenotype PGM$_1$ (8-a2) was excluded from the analyses of the data

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