THE DISTRIBUTION OF THE PHOSPHOGLUCOMUTASE-I (PGM₁) SUBTYPES IN JAPANESE¹

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Summary PGM₁ subtypes in Japanese were examined by thin-layer polyacrylamide gel isoelectrofocusing (pH 5-7) using fresh hemolyzates from 160 unrelated adults and members of 16 families living in Tokyo. The results were consistent with the model proposed in Europeans in that ten PGM₁ subtypes are determined by 4 codominant alleles. The allele frequencies estimated in Japanese are as follows. \( PGM₁^{1+} : 0.6154 \), \( PGM₁^{1-} : 0.0833 \), \( PGM₁^{2+} : 0.2340 \), \( PGM₁^{2-} : 0.0673 \).

Human phosphoglucomutase-I (PGM₁) is a well known genetic marker showing polymorphism; three common phenotypes, PGM₁ 1, PGM₁ 2-1 and PGM₁ 2 are determined by a pair of autosomal alleles: \( PGM₁^1 \) and \( PGM₁^2 \) (Spencer et al., 1964). Recently, use of thin-layer polyacrylamide gel isoelectrofocusing has led to the detection of two subtypes of each of the isozyme products of \( PGM₁^1 \) and \( PGM₁^2 \) (Bark et al., 1976; Kühnl et al., 1977; Sutton et al., 1978). Thus, it now appears that the PGM₁ polymorphism comprises as many as 10 phenotypes determined by four alleles. To date, the data of the distribution of PGM₁ subtypes are confined to European populations. In this paper we report the data of the distribution of PGM₁ subtypes in a Japanese population.

MATERIALS AND METHODS

Fresh hemolyzates were obtained from 160 unrelated blood donors living in Tokyo. Moreover, samples from 16 pairs of twins and their parents were examined. Hemolysates were usually prepared from washed, packed red cells stored at \(-20^\circ\text{C}\) with an equal volume of 3% glycerol-saline solution.

Samples were screened firstly by starch gel electrophoresis using the method essentially following Spencer et al. (1964), and then subjected to the thin-layer polyacrylamide electrofocusing at pH 5-7. The isoelectrofocusing conditions were similar to those reported by Bark et al. (1976) with slight modifications.

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The slab gel (16 x 10 x 0.1 cm) was made by mixing the following solutions and polymerization for overnight under a fluorescent lamp.

3 ml acrylamide solution (29.1 g/100 ml H₂O)
3 ml NN'-methylenebisacrylamide solution (0.9 g/100 ml H₂O)
10 ml sucrose solution (25 g/100 ml H₂O)
1.5 ml ampholine solution (LKB, pH 5-7)
0.2 ml riboflavin solution (4 mg/100 ml H₂O)

Isoelectrofocusing was carried out using the apparatus manufactured by Joko Sangyo Co., Tokyo, in which water at 4°C was circulating. Samples (16 per gel in standard run) were applied using 3 x 5 mm pieces of Whatman 3MM filter paper at 3 cm from the anodal edge of the gel. Starting from an initial voltage of 300V, the final voltage of 1,000V was attained in about 70 min, and thereafter the run was continued for about 5 hr. PGM staining was carried out by the method originally described (Spencer et al., 1964) with slight modifications, using the increased amount of G-1-P (BDH), NADP (Oriental) and G-6PD (Oriental) in the reaction mixture. The staining mixture was applied as agar overlay and incubated at 37°C in the dark for 30-60 min. The PGM₁ isozymes appeared on the gel in about 30 min as sharp bands. The PGM₂ isozymes do not form sharp bands in the experimental conditions mentioned, and therefore not confused with the PGM₁ isozymes. For preservation of the gel, it was washed in cooled water to remove excess reagents and then dried on the glass plate.

RESULTS AND DISCUSSION

Excluding those samples with variant PGM₁ phenotypes (PGM₁ 7-1, 6-1 etc.) detected by starch gel electrophoresis, 156 samples with common PGM₁ phenotypes were subjected to isoelectrofocusing. Samples of the classical PGM₁ homozygotes, PGM₁ 1 and PGM₁ 2, were found to have either a single major PGM₁ band or two PGM₁ bands, confirming the existence of two subtypes for each of the PGM₁ 1 and PGM₁ 2 isozymes (Fig. 1). Each PGM₁ isozyme was accompanied by an anodal minor band. As for PGM₁ 2 isozymes, however, the staining intensity of this minor band is almost as strong as that of the major band.

Though different notations are proposed by different investigators, we prefer to use + − symbols to distinguish subtypes. The patterns of the possible 10 PGM₁ phenotypes are diagrammatically shown in Fig. 2. All but one (PGM₁ 1−) phenotypes were found as shown in Table 1. Three phenotypes, PGM₁ 1+, 1+1−, 2+1+, are exceedingly common, with phenotypic frequency of 39.10%, 11.54%, 26.28%, respectively. Allele frequencies were estimated as follows: PGM₁ 1+: 0.6154, PGM₁ 1−: 0.0833, PGM₁ 2+: 0.2340, PGM₁ 2−: 0.0673. The observed numbers of phenotypes were in good agreement with the numbers expected under the equilibrium condition (χ²=5.53, 6df, 0.50<p<0.70).

The results of PGM₁ subtyping of samples from 16 twin pairs and their parents