

1-1-1998

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PERÇİN, E. Ferda and GÖZE, A. İsmihan (1998) "A New Phenotype in the PhosphoGLUcomutase1 (PGM1) System, PGM1*W32," *Turkish Journal of Medical Sciences*: Vol. 28: No. 6, Article 5. Available at: <https://journals.tubitak.gov.tr/medical/vol28/iss6/5>

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A New Phenotype in the Phosphoglucomutase 1 (PGM1) System, PGM1*W32

Received: October 01, 1996

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Abstract: Red cell phosphoglucomutase 1 (PGM1) enzyme, which is a phosphotransferase, has 3 common and 31 rare phenotypes.

In this study, we have identified a new phenotype for this enzyme which has not been reported previously using cellulose

acetate electrophoresis technique. This phenotype has also been identified for the first time for Turkish population and called PGM1*W32.

Key Words: PGM1, phenotype, rarephenotype.

Introduction

In view of its central role in glycolysis and gluconeogenesis and its polymorphic variability, the phosphoglucomutase 1 (PGM1) gene in man has been the target of protein structural studies and genetic analysis for more than 25 years.

Phosphoglucomutase (PGM: E.C.2.7.5.1) plays an important role in carbohydrate metabolism. It is a phosphotransferase which catalyses the transfer of phosphate group between the first and sixth positions of glucose (glucose -1- phosphate ↔ glucose -6- phosphate). PGM occurs in all human tissues and is controlled by four autosomal loci, i.e., PGM1, PGM2, PGM3 and PGM4. Each locus determines on characteristic set of 2 or 3 isozymes which migrate anodally in the order PGM1<PGM2<PGM3 [1-5]. The polymorphism at the PGM1 locus in human was first revealed in 1964 [1]. Since then, extensive studies showed that two codominant alleles, PGM1*1 and PGM1*2 which are expressed as phenotypic 1,1, 2-1, 2-2, are present on locus PGM1 in all populations, as well as many rare alleles. The workshop, held in West Germany in 1985, recommended a new nomenclature: PGM1*W1-W30 in order to avoid misunderstanding. In accordance with the decision held in the workshop it is suggested that the whole family should be analyzed and two different techniques should be applied [6]. A new phenotype called PGM1*W31 was then identified [7].

In summary, to date, 3 common and 31 rare phenotypes for PGM1 have been identified.

In this study, we have identified a new phenotype of PGM1, which we have called PGM1*W32, using cellulose acetate electrophoresis. This phenotype is also new for Turkish population.

Materials and Methods

Venous blood was drawn from diabetic patients and a family which we have called "Y" family. 1.6 ml. blood was placed in tubes containing 0.4 ml. sodium citrate solutions. The plasma was then separated, saponin added for hemolysis and frozen at -20°C. The cellulose acetate electrophoresis method developed by Grunbaum was used for the determination of PGM1 isoenzymes [8].

Results

Commonly observed PGM1 1-1 and PGM1 2-1, which are PGM1*1/PGM1*1 and PGM1*2/PGM1*1 genotypes respectively, and new phenotypes of PGM1 have been shown in Fig.1 and diagrammatic appearance in Fig.2. When the experiment was repeated in case of any technical error, the same phenotype was observed (Fig.3). The same phenotype was seen in no member of "Y" family as the other diabetic individuals analyzed. This pedigree pattern is shown in Fig.4. Phenotypes of proband's children were determined to be PGM1 2-1 by cellulose acetate electrophoresis. It has been noticed that the phenotype determined in our investigation was not similar to 3 common and 31 rare phenotypes of PGM1. It should also be noted that, although individual with new

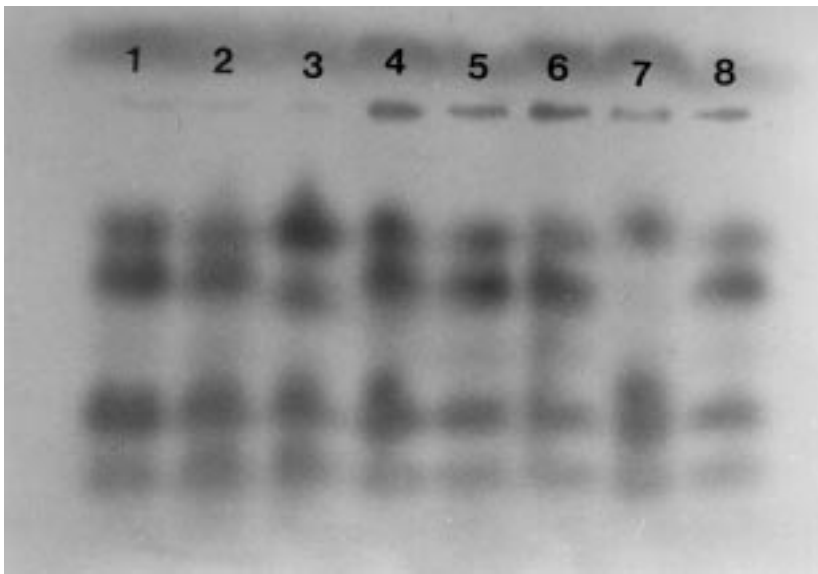


Figure 1. PGM1 phenotyping on cellulose acetate membrane (Each phenotype is for a different diabetic individual)
 Lane 1: PGM1 1-1, Lane 2: PGM1 1-1, Lane 3: PGM1 1-1, Lane 4: PGM1 1-1, Lane 5: PGM1 2-1, Lane 6: PGM1 2-1, Lane 7: PGM1 new, Lane 8: PGM1 2-1.

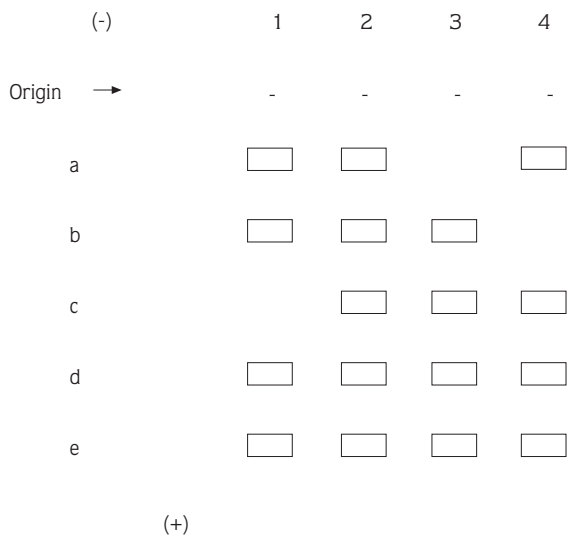


Figure 2. Diagram of the PGM1 enzyme phenotypes (According to electrophoresis, patterns of band were shown as a,b,c,d,e)
 Lane 1: PGM1 1-1, Lane 2: PGM1 2-1, Lane 3: PGM1 2-2, Lane 4: New Phenotype

phenotype that we identified was diabetic, we have not found any statistical correlation with diabetes for this phenotype. According to the suggestion, given in the "Report on the International PGM1-Workshop" the new phenotype was named as PGM1*W32.

Discussion

In accordance with the decision held in the workshop, it is claimed that the whole family, of the individual who

is determined as a new phenotype, should be analyzed by applying two different techniques. Because to establish a new phenotype by applying starch gel, cellulose acetate electrophoresis techniques is much more difficult than the methods as isoelectric focusing (IEF) system which gives out a number of band samples. As a matter of fact while the researchers finding at PGM1*W31 couldn't find a new phenotype by applying cellulose acetate electrophoresis technique, they managed to establish this fact in IEF on polyacrilamide gel [7]. We have no doubt that a phenotype which can be seen even by applying cellulose acetate electrophoresis technique can be established by the techniques which can show more bands. However, we couldn't apply the mentioned method by the possibilities our hold.

The proband's (II-3) phenotype was considered to be homozygote because mode of inheritance of this enzyme was known to be autosomal codominant. The new phenotype found in proband might have resulted from new mutation in proband or from her mother who is not alive. In contrast to band pattern seen in proband, her children have 2-1 phenotype in which second band is present. Therefore they might have inherited this second band from their father.

In investigation mode in Turkey on this subject no rare phenotype has been reported [4,9-12]. According to Canlı, one PGM1 6-1 phenotype has been encountered in 243 blood samples obtained from the Turkish-Cypriot population living in England [9]. However, the phenotype we found is not consistent with this sample. According to literature the most frequently observed rare phenotypes

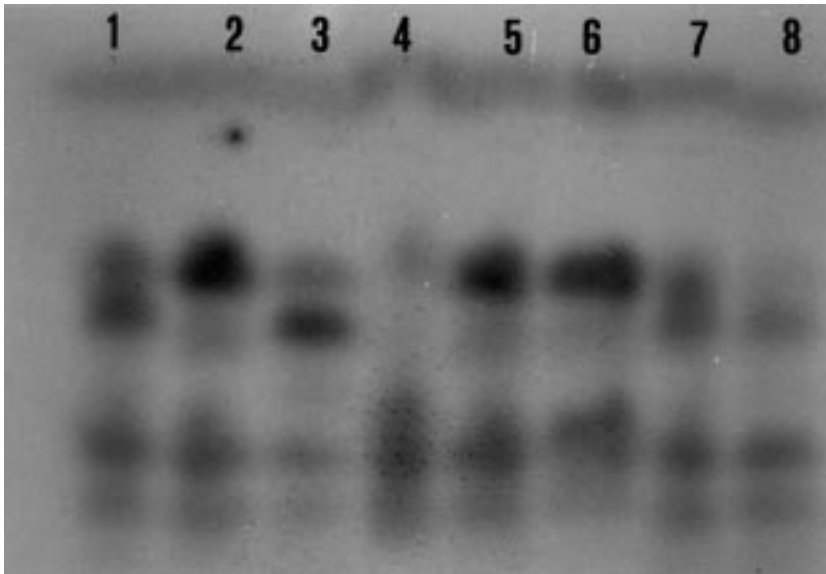


Figure 3. PGM 1 phenotyping on cellulose acetate membrane (Each phenotype is for a different diabetic individual)
 Lane 1: PGM1 1-1, Lane 2: PGM1 1-1, Lane 3: PGM1 2-1, Lane 4: PGM1 new, Lane 5: PGM1 1-1, Lane 6: PGM1 1-1, Lane 7: PGM1 1-1, Lane 8: PGM1 1-1.

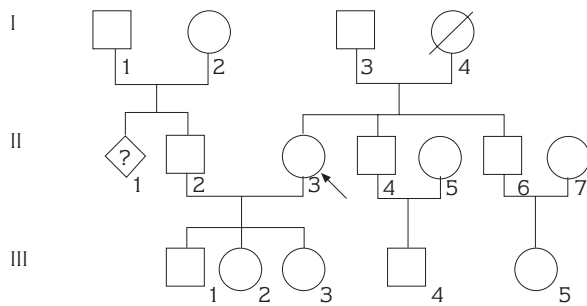


Figure 4. I-3: 2-2, II-3: new, II-4: 2-1, II-6: 2-1, III-1: 2-1, III-2: 2-1, III-3: 2-1, III-4: 2-2, III-5: 1-1.

of PGM1 were found to be PGM1 3-1 and PGM1 7-1 which were reported to belong to Asians in more than 50% of the cases [8, 13].

Although, the phenotypes we found belonged to a diabetic individual, it was not statistically related with diabetes in an investigation carried out with 50 people with diabetes [14]. In another study, 150 diabetic patients displayed no other rare phenotypes (unpublished result). To date, no such finding was reported in any other study carried out with diabetic patients [15-17].

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