

Silent Alleles

Contrary Homozygosity of The PGM₁ System in Disputed Paternity

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INTRODUCTION

Since the original discovery of the polymorphism of phosphoglucomutase in human red cells /Spencer et al. 1964/, a number of investigators and population studies of the enzyme have been described. Isoelectric focusing either in polyacrylamide or agarose gels has become the method of choice for subtyping of the PGM isoenzymes determined by the PGM₁ locus /Bark et al. 1976; Kühnl et al. 1977; Sutton and Burgess¹1978/.

During the determination of blood groups in forensic cases of disputed paternity in the last year 1986 in two cases an incompatibility between a putative father and a child in PGM₁ system was observed.

METHODS

All samples of blood have been investigated routinely in following genetic marker systems such as: ABO, Rh, MNSs, Kk, P₁, Hp, Gm 1, 2, Gc, C3, AcP, PGM₁, EsD, GLO and HLA-ABC. The PGM₁ phenotypes were tested by use starch gel electrophoresis and by means of isoelectric focusing /IEF/.

RESULTS

The following results was observed:

First case

Putative father: O, MSs, Rh: CcDee, kk, P₁+, Hp 2-1, Gm 1, 2, Gc 1, C3 FS, AcP B, EsD 1, GLO 2, PGM₁ 2, HLA-A3, 11, B35, 44/12, w4, w6, Cw4, w7

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Proband: O, MSSs, Rh:CCDee, kk, P₁+, Hp 2-2, Gm 1, -2, Gc 1, C3 FS, AcP B, EsD 2-1, GLO 2-1, PGM₁ 1, HLA-A3, B14, 44/12/, w4, w6, Cw7

Mother of proband: O, MNSS, Rh:CCDee, kk, P₁-, Hp 2-2, Gm 1, -2, Gc 1, C3 FS, AcP B, EsD 2, GLO 1, PGM₁ 1, HLA-A2, 3, B14, w62/15/, w6, Cw3

Second case

Putative father: O, MNSSs, Rh:CcDee, kk P₁+, Hp 1-1, Gm 1, 2, Gc 1, C3 FS, AcP B, EsD 1, GLO 2, PGM₁ 1, HLA-A2, 24/9/, Bw 56/22/, w62/15/, w6, Cw3

Proband: A₂, MSSs, Rh: CcDee, kk, P₁+, Hp 1-1, Gm 1, 2, Gc 2-1, C3 S, AcP B, EsD 1, GLO 2-1, PGM₁ 2, HLA-A11, 24/9/, B13, w56/22/, w4, w6, Cw3, w6

Mother of proband: A₂, MSSs, Rh:CcDee, kk, P₁+, Hp 1-1, Gm 1, -2, Gc 2-1, C3 FS, AcP BA, EsD 1, GLO 2-1, PGM₁ 2, HLA-A1, 11, B13, w4, Cw6

In these findings the putative fathers ought to be excluded. For this reason the PGM₁ subtypes were examined by means of IEF. The results were as follows:

	first case	second case
putative father	2-	1+ 2-
proband	1+ 2-	2-
mother of proband	1+	2- 2+

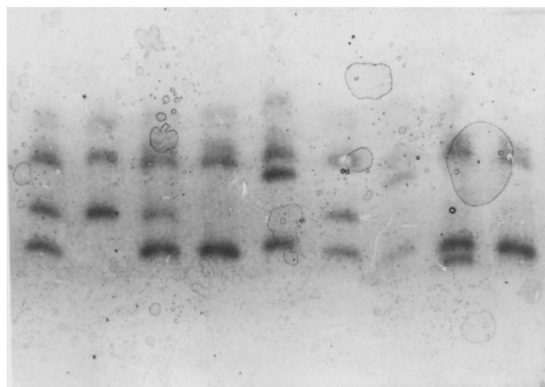


Fig. 1. Photograph of PGM₁ subtypes determined by IEF on 0.5mm polyacrylamide gel /pH 5-7/. Samples from left to right: 1+2-, putative father 2-, child 1+2-, mother 1+, 1+2+, 1+2-, 1+2+, 1+1-, 1+ /first case/

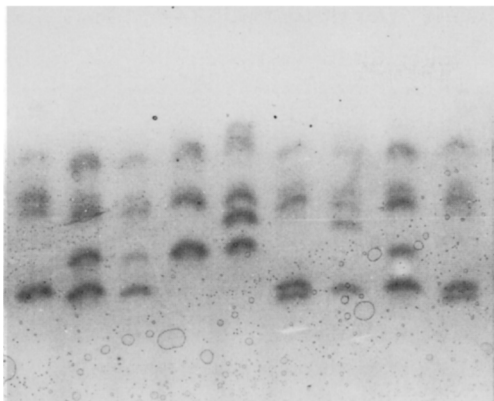


Fig. 2. Photograph of PGM_1 subtypes determined by IEF on 0.5mm polyacrylamide gel /pH 5-7/. Samples from left to right: 1+, 1+2-, putative father 1+2-, child 2-, mother 2-2+, 1+1-, 1+2+, 1+2-, 1+1- /second case/

Polyacrylamide gel isoelectric focusing revealed the existence of weak bands in the proband /first case/ and in putative father /second case/ patterns corresponding to isoenzyme 2- /Fig. 1 and Fig. 2/. In these findings the putative fathers were not excluded.

The results suggest that not only typing by starch gel electrophoresis but also subtyping by isoelectric focusing should be performed, especially in forensic investigations such as testing for paternity.

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