

PGD Duisburg: A new Variant of 6-Phosphogluconate Dehydrogenase

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INTRODUCTION

Human 6-phosphogluconate dehydrogenase (PGD-E.C.1.1.1.44) is an important metabolic enzyme in the hexose monophosphate shunt pathway. Genetically determined polymorphism has been described 1963 by Fildes and Parr using starch gel electrophoresis. In addition to the two common phenotypes, PGD A and PGD AB, a number of electrophoretically fast and slow moving rare variants has been observed (Parr 1966; Davidson 1967; Tariverdian et al. 1970; Blake et al. 1974; Spielmann and Kühnl 1982). Inherited quantitative variations of this enzyme have also been reported (Parr and Fitch 1967).

In this paper we describe a new PGD variant along with population data from Germany. This variant which has been tentatively named PGD Duisburg can be differentiated by its electrophoretic mobility and banding intensity. Family studies confirmed its transmission.

MATERIALS and METHODS

The study includes 2020 unrelated individuals from Germany. Hemolysates were prepared by sonification of twice-washed erythrocytes.

Horizontal starch gel electrophoresis was carried out overnight for 15 h at a voltage gradient of 10 V/cm with a 0.1 M phosphate buffer at pH 7.0. NADP was routinely added to the bridge and gel buffer with a final concentration of 2 mg/dl. PGD isozyme bands were visualized on the gel, using the staining agar overlay method described by Harris and Hopkinson (1976).

RESULTS and DISCUSSION

Figure 1 represents the banding patterns of five different PGD phenotypes after starch gel electrophoresis. The newly observed variant PGD Duisburg (PGD D) moves electrophoretically faster than the common PGD A but slower than PGD Richmond (PGD R). Phenotype PGD AD shows a close triplet pattern. The anodal band of this triplet has a similar position as the

intermediate band of PGD R. The slowest band is an A band, and the middle band is symmetrically placed and has substantially greater intensity than the bands on either side of it. The triplet can be observed only with a high resolution of bands otherwise it looks like a single A band. The banding pattern is not affected by either NADP or dithiothreitol and it is also thermostable. According to its thermostability and electrophoretic pattern this new variant is different to PGD Friendship. Another symmetrically triplet pattern is shown by the faster moving variant phenotype PGD AR and the slower moving PGD AH (Hackney).

Figure 2 gives a diagrammatic representation of the observed PGD phenotypes.

The pedigree of family Ko. is shown in Figure 3. It demonstrates the transmission of the allele PGD *D from the father to one of his four children. Three of these children are homozygote PGD A. There was no possibility to determine

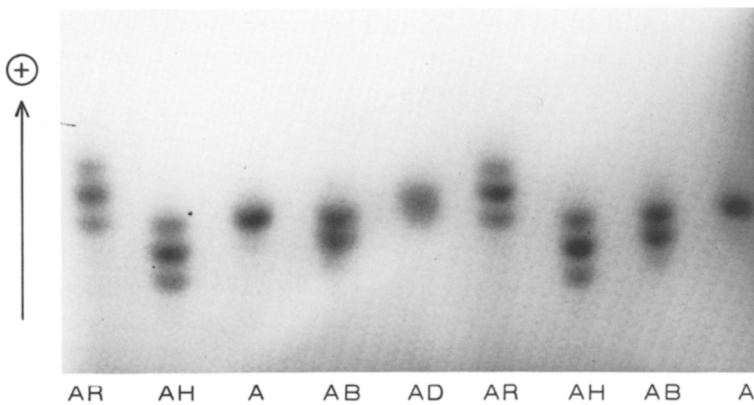


Fig. 1. Banding patterns of PGD phenotypes after starch gel electrophoresis of hemolysates

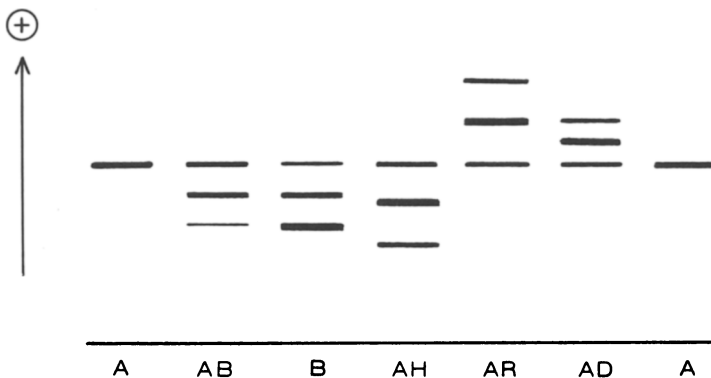
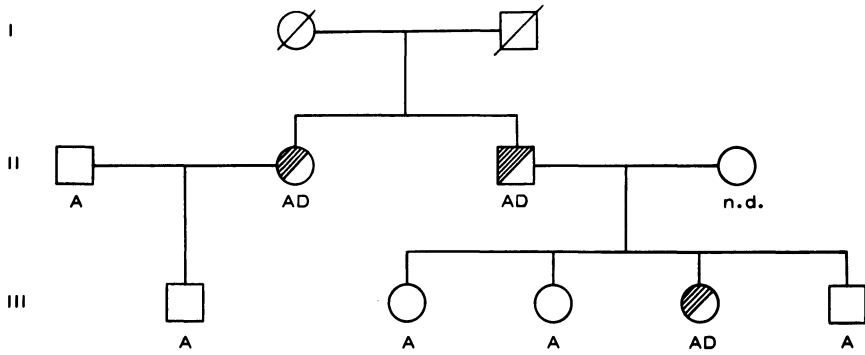


Fig. 2. Schematic representation of the observed PGD phenotypes

the mother's phenotype. The variant PGD Duisburg, which indicates the father's birthplace, was also found in his sister by paternity testing.



n.d. = not determined

Fig. 3. Pedigree of family Ko. Individuals with the variant PGD Duisburg are indicated with half black symbols

Table 1. Distribution of PGD phenotypes and alleles in a sample from Germany

Phenotypes	Observed		Expected		Allele frequencies
	n	%	n	%	
PGD A	1934	95.74	1934.86	95.83	PGD*A = 0.9787 PGD*B = 0.0200 PGD*Var= 0.0013
AB	81	4.01	79.08	3.92	
B	1	0.25	5.14	0.25	
AR	2				
AH	1				
AD	1				
Total	2020	100.00	2019.08	100.00	

$$\sum \chi^2 = 0.0508; df = 1; P > 0.20$$

The distribution of PGD phenotypes and alleles in a sample from Germany is shown in Table 1. In addition to the two common phenotypes the rare type PGD B as well as the variant phenotypes PGD AR, PGD AH and PGD AD were found in this study. Observed and expected distribution are in good agreement with the Hardy-Weinberg equilibrium. The allele frequencies correspond to those found in other European populations (Prokop and Göhler 1986). Despite the extensive studies carried out on the PGD polymorphism in many different populations, to our knowledge PGD Duisburg is a further rare variant in this system.

ACKNOWLEDGEMENT. We would like to thank Dr. P. Kühnl, Institut für Immunhämatologie der Universität Frankfurt, for confirming the new variant phenotype identified in this study.

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