PGM1 Subtyping by Isoelectric Focusing (IEF) in Parentage Testing in South African (SA) Populations

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INTRODUCTION

PGM1 is an ubiquitous enzyme which is coded for by a gene situated on chromosome 1p. PGM1 reversibly catalyses the transfer of phosphate from the first to the sixth position of glucose in the glycolytic pathway. The polymorphism of PGM1 was first demonstrated by conventional starch gel electrophoresis by Spencer et al in 1964. Using IEF, later studies showed more complex band patterns due to the presence of four common alleles at the PGM1 locus, giving rise to ten possible phenotypes (Bark et al. 1976; Sutton and Burgess 1978). PGM1 is one of thirteen genetic marker systems currently used in disputed paternity testing in this laboratory. The aim of this study was to assess the usefulness of PGM1 subtyping by IEF in disputed parentage testing and compare the results with those obtained using starch gel electrophoresis.

MATERIALS AND METHODS

Acid citrate dextrose anticoagulated blood samples were obtained from 638 mother-child-alleged father trios, from SA Caucasoids, Cape Coloureds and SA Negroes (Xhosa).

Haemolysates were prepared from washed packed cells treated with 10% Triton X-100 and stored at -70^{0}C. Polyacrylamide gels were prepared at a concentration of T=5% and C=3% with an ampholyte range pH 5.0 - 7.0. Five ul of haemolysate were applied on filter paper 2cm anodally. The anolyte was 1M phosphoric acid. The catholyte was 1M sodium hydroxide. IEF was performed at 1200V, 15mA, 20W at 10^{0}C for 180 minutes. Enzyme activity was revealed using the electron transfer stain, as described by Sutton and Burgess (1978).

Gene frequencies were established by direct counting.
The observed power of exclusion (PE) was calculated by counting the number of men excluded by the PGM1 system, out of the total number of men excluded, using thirteen systems.

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RESULTS

Table 1. Observed and expected PGM1 phenotype frequencies

	SA Caucasoids			Cape Coloureds			SA Negroes (Xhosa)		
Pheno-types	Obs.	Exp.	X2	Obs.	Exp.	X2	Obs.	Exp.	X2
1A	100	102.47	0,06	242	231.47	0.48	174	176.91	0.05
1B	1	3.44	1.73	13	15.50	0.40	12	16.61	1.28
2A	10	8.55	0.25	12	11.29	0.05	18	16.04	0.24
2B	2	1.01	0.97	2	.73	2.20	0	0.21	0.21
1A1B	44	37.55	1.11	116	119.80	0.12	119	108.41	1.03
2A1A	60	59.19	0.01	93	102.24	0.84	103	106.55	0.12
2B1A	18	20.35	0.27	18	26.02	2.47	11	12.20	0.12
2A1B	8	10.84	0.75	32	26.46	1.16	31	32.65	0.08
2B1B	5	3.73	0.43	10	6.74	1.58	4	3.74	0.02
2A2B	5	5.88	0.13	8	5.75	0.88	5	3.67	0.48
Total	253	253.01	5.71	546	546.00	10.18	477	476.99	3.63

Table 1 shows the observed and expected phenotype frequencies for the three populations studied. The gene frequencies for the PGM1 alleles in SA Caucasoids, Cape Coloureds and SA Negroes are shown in Table 2. The observed and expected values of the phenotypes within each population were in accordance with Hardy-Weinberg equilibrium.

Table 2. Distribution of PGM1 gene frequencies

	SA Caucasoids n = 253	Cape Coloureds n = 546	SA Negroes (Xhosa) n = 477
1A	0.6364	0.6511	0.6090
1B	0.1166	0.1685	0.1866
2A	0.1838	0.1438	0.1834
2B	0.0632	0.0366	0.0210

Figure 1 shows the comparison between the power of exclusion in the various South African populations using PGM1 typing by either starch gel electrophoresis or IEF.

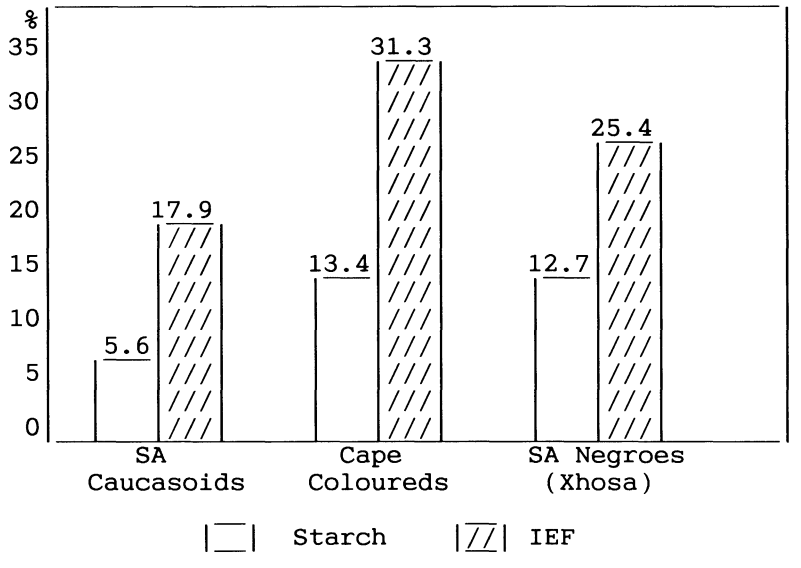

Figure 1. Comparison of the power of exclusion (PE) of PGM1 using starch gel electrophoresis and IEF

CONCLUSION

In view of the increased power of exclusion of PGM1 detected using IEF, this method is more useful than starch gel electrophoresis especially in the Cape Coloureds. Therefore PGM1 detected using IEF is recommended as an additional system for disputed parentage testing.

REFERENCES

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