4.3 *Practical Application*

Parentage Testing Using DBP Subtyping in South African (SA) Populations

W. Petersen, T. Schlaphoff, R. Martell and E. du Toit

Provincial Laboratory for Tissue Immunology, Cape Town, South Africa

INTRODUCTION

The vitamin D binding protein (DBP) of human plasma, also referred to as group specific component (GC), was first described by Hirschfeld in 1959. One of its major functions is to act as a transport protein for vitamin D-3 and its naturally occurring derivatives.

Family studies have confirmed that the DBP phenotypes are determined by a pair of autosomal co-dominant alleles. By using an isoelectric focusing (IEF) subtyping technique Constans and Viau (1977), distinguished six common phenotypes, determined by 3 alleles, namely DBP*1S, DBP*1F and DBP*2. Using various techniques, more than 120 allelic variants of the DBP system have been identified to date.

MATERIALS AND METHODS

This study analysed the results of serum samples collected from 238 SA Caucasoids, 515 Cape Coloureds and 465 SA Negroes (Xhosa), all of whom were the parents in disputed paternity cases. Samples were frozen at -70°C or -20°C until used.

The technique involved IEF subtyping in ultrathin polyacrylamide gel using a narrow ampholyte range pH 4.5-5.4. Visualisation of the bands was by immunofixation, using a monospecific antiserum on cellulose acetate followed by staining with Coomassie Brilliant Blue. An agarose gel using a barbital buffer was used to confirm the DBP*AM and DBP*AB variants. For the purpose of paternity testing the old nomenclature, DBP*AM and DBP*AB, was used, since it was found unneccessary to further identify them (personal correspondence, Cleve, H).

The frequencies of the DBP alleles were determined by direct counting. The power of exclusion (PE) was calculated by dividing the number of cases with a DBP exclusion by the total number of excluded cases, using 14 genetic systems, namely, HLA, ABO, RH, MNSs, FY, KEL, ACP1, ESD, PGM1(IEF), CA2, GLO1,

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DBP(IEF), HP and BF. The cumulative power of exclusion (CPE) using the 14 genetic systems was established by using the formula:

$$CPE = 1-(1-PE_1)(1-PE_2)...(1-PE_n).$$

RESULTS

Table 1. Observed and expected DBP phenotype frequencies

	SA Caucasoids			Cape Coloureds			SA Negroes (Xhosa)		
Pheno-types	Obs.	Exp.	X^2	Obs.	Exp.	X^2	Obs.	Exp.	X^2
1S	68	68.84	0.01	44	39.72	0.58	0	2.56	2.56
1F	10	7.24	1.05	153	142.06	0.84	312	313.81	0.01
2	18	19.14	0.07	17	13.27	1.05	3	1.69	1.02
AM	0	0	0	0	0.26	0.26	0	0.26	0.26
AB	0	0	0	0	0.02	0.02	0	0.11	0.11
Var	0	0	0	0	0.04	0.04	0	0.01	0.01
1S-1F	42	44.65	0.11	140	150.22	0.70	60	56.69	0.19
2-1S	78	72.60	0.40	50	45.82	0.38	3	4.15	0.32
1S-AM	0	0	0	4	6.38	0.89	4	1.63	1.45
1S-AB	0	0	0	1	1.66	0.26	1	1.04	0
1S-Var	0	1.08	1.08	3	2.52	0.09	1	0.37	1.07
2-1F	20	23.54	0.53	75	86.66	1.57	46	45.99	0.
1F-AM	0	0	0	14	12.06	0.31	17	18.03	0.06
1F-AB	0	0	0	3	3.14	0.01	13	11.54	0.19
1F-Var	1	0.35	1.21	3	4.76	0.65	4	4.13	0.00
2-AM	0	0	0	3	3.68	0.13	1	1.32	0.08
2-AB	0	0	0	2	0.96	1.13	0	0.85	0.85
2-Var	1	0.57	0.32	1	1.45	0.14	0	0.30	0.30
AM-AB	0	0	0	0	0.13	0.13	0	0.33	0.33
AM-Var	0	0	0	2	0.20	0.20	0	0.12	0.12
AB-Var	0	0	0	0	0.05	0.05	0	0.08	0.08
Total	238	238.01	4.78	515	515.06	9.43	465	465.01	9.01

Table 1 shows the observed and expected DBP phenotype frequencies in the three populations studied. As shown in Table 2 DBP*1S is more common in the SA Caucasoids and DBP*1F is more common in the SA Negroes (Xhosa). The Cape Coloureds, who are of mixed ancestry, have DBP allele frequencies which are intermediate between those of the SA Caucasoids and SA Negroes. According to this study, some of the DBP alleles are population specific, for example the DBP*AM and DBP*AB alleles appear to be Negroid genes. These populations are in Hardy-Weinberg equilibrium, as estimated from these gene frequencies.

Table 2. Distribution of DBP gene frequencies

	SA Caucasoids n = 238	Cape Coloureds n = 515	SA Negroes (Xhosa) n = 465
1S	0.5378	0.2777	0.0742
1F	0.1744	0.5252	0.8215
2	0.2836	0.1602	0.0602
AM	0.0	0.0223	0.0236
AB	0.0	0.0058	0.0151
variants	0.0042	0.0088	0.0054

The results in Table 3 indicate the usefulness of including DBP subtyping by IEF, in paternity testing, especially in the Cape Coloureds.

Table 3. Power of exclusion (PE) using the DBP system

Exclusions	SA Caucasoids	Cape Coloureds	SA Negroes (Xhosa)
DBP	6	37	19
Total*	28	115	122
PE%	21.4	32.2	15.6

*using 14 genetic systems.

CONCLUSION

Vitamin D binding protein subtyped by isoelectric focusing, is useful in conjunction with other genetic systems for excluding a falsely accused father of parentage especially in the Cape Coloured population.

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