Bioassay of kinship using VNTR alleles

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INTRODUCTION

Much attention has been given to forensic statistics. Focus has been on matching criteria, on binning procedures and likelihood ratios, and on VNTR genotype distribution in forensic populations (e.g. Lander 1989, Cohen 1990, Chakraborty 1991, Lewontin and Hartl 1991, Evett and Pinchin 1991, Nichols and Balding, 1991). Newton Morton deserves much credit for his valuable contributions in the field (see e.g. Morton 1992). Recently he and his colleagues demonstrated how probability calculations in forensic identification may be reliably performed if due care is given in particular to kinship (Morton et al 1993).

Kinship (F) is the probability that two alleles are identical by descent. In modern society, F in matings is ordinarily very small - and less than 1 per cent. Ordinarily allele frequencies are so high that estimates of kinship in matings would require extremely large materials. It struck us - however - that given a maximal electrophoretic exploitation of VNTR fragment length polymorphisms, allele frequencies might be brought down to a level which should allow estimates of kinship in such pairwise relationships.
The aim of our study was to establish a procedure for bioassay of kinship suitable for evaluating pairwise relationships like matings, alleged fathers, and suspects in criminal cases.

MATERIAL AND METHODS

The material we had at hand was 1403 consecutive paternity cases, all typed in the VNTR loci D2S44, D7S21, D7S22, D12S11, and D14S13.
We sorted three different kinds of pairs: 1052 between-case female/male pairs, representing the general paternity case population, 1403 within-case female/male pairs representing paternity case matings, and 185 within case male/male pairs representing the relationship between males involved in the same paternity case.
Only intragel comparisons between individuals not more than 2 lanes apart were included.
The matching criterium chosen was a perfect visual match - that is when an experienced typer decides that the alleles might well be identical. In our experience this matching criterium equals a sliding window of 0.2-0.3 mm.

RESULTS AND DISCUSSION

Fig 1 illustrates the allele discrimination level obtained. It shows 5 pairs originally typed either as matches or near matches - within 1 mm apart. Pairs number 1 and 5 are matches, number 3 and 4 are non-matches about 0.5mm apart, while nomber 2 - after some hesitation - is typed as a match.

Fig 1

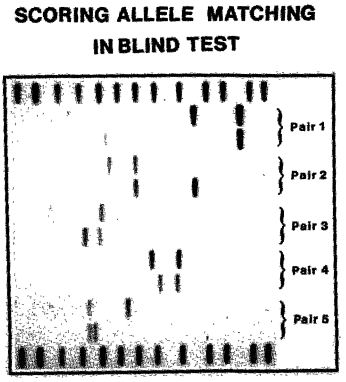

Table I TEST FOR TYPING BIAS

	ORIGINAL type distribution		BLIND TEST type distribution	
	match	near-match *	match	near-match *
between case pairs	50	50	51	49
within case pairs	50	50	52	48
mother/child pairs	100		98	2

*· less than 1mm migration distance, but not a perfect visual match

Since the original typing procedure did not allow blind typing, a blind test was performed which involved more than 10 per cent of all matches as well as a similar number of near-matches and obligate mother-child pair matches.
This test demonstrated that there was no systematic typing bias in the original typing, and that very few true matches were lost (Table I).

Table II MATCHING OF ALLELES IN A PATERNITY CASE MATERIAL
Comparison between 1052 between-case and 1588 within-case pairs of individuals, using 5 highly polymorphic VNTR probes

TYPE OF PAIR	Number of pairs	Allele compar.	Allele matches Number	Percent	KINSHIP %
Between-case	1052	21040	307	1.46	
Within-case					
female/male	1403	28060	530	1.89	0.436
male/male	185	3700	101	2.73	1.289

Table II shows the main results of our study. The between-case allele matching frequency of 1.46 per cent is far below the homozygote frequency of 5-10 per cent reported when traditional matching criteria are used (See e.g. Morton et al 1993). It illustrates that the present procedure exploits the hyperpolymorphism extensively.
The difference in allele matching frequency between between-case and within case female/male pairs is statistically significant. Kinship in paternity case matings (0.436%) is in accordance with kinship levels in Norway estimated by genealogic methods (Gedde-Dahl 1973).
The allele matching frequency in pairs of males involved in the same paternity case is 2.7 per cent, nearly twice that of between case pairs. This means that the kinship factor is of about the same size as is the allele frequency. Kinship is estimated at 1.3 per cent, a surprisingly high value. Data not shown here indicate that there is a 5-10 per cent proportion of closely related males between men involved in the same case.
The results demonstrate that bioassay of kinship by the procedure described is an effective tool to estimate kinship in pairwise relationships. Kinship levels in paternity case parties should be taken into account in paternity statistics. Studies in pairs of suspects in forensic casework might be of interest for forensic statistics as well.

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