PCR POLYMORPHISMS COMPARED TO OTHERS GENETIC MARKERS IN THE PATERNITY TESTING

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INTRODUCTION

Paternity testing is an important aspect of the Forensic Laboratory in Spain since the reform of the civil code of 1981 about filiation. Peculiarities of the catalan civil law in paternity allowed the Forensic Hemogenetics Investigation Group of the School of Medicine of the Barcelona University to became one of the pioneers in this field in Spain. Ours was the first forensic laboratory in Spain that employed the HLA polymorphism to the paternity testing (1976).

The use of several techniques improves the confiance of the diagnostic so, by employing simultaneously red blood cell groups (RBC), plasma protein systems (PP), red cell enzyme systems (EE) and Human Leucocyte Antigens groups (HLA), a high standard of reliability was achieved in paternity exclusion as well as in inclusion.

The last biotechnological advances allowed to apply methods of DNA polymorphism in the field of the Forensic Medicine. The first DNA polymorphisms applied to Forensic Medicine, were the RFLP's (RESTRICTION FRAGMENT LENGHT POLYMORPHISM). Soon, several problems involved with allelic identification in population studies were resolved by using techniques based on POLYMERASE CHAIN REACTION (PCR). The PCR offered obvious advantages in paternity testing.

In 1990 we included the PCR method in our work in paternity testing. Presently we study five loci: D1S80 (pMCT118), 3'Apo B, D17S5 (YNZ22), HLA DQα and ACTBP2 (SE33). D1S80, D17S5, 3'ApoB and ACTBP2 loci, are VNTR polymorphisms (Variable Number of Tandem Repeats). The HLA DQα locus presents an ASO polymorphism (Allele Specific Oligonucleotide). PCR polymorphisms increase the information in paternity testing because of their variability, thus, reliability is improved.

In this work we have calculated the statistical parameters of the five polymorphisms studied by PCR, and we have compared them to the rest of the classical genetic markers. This allows to know accurately their efficiency in paternity testing.

METHODS

After having carried out the research in Catalonian population and getting gene frequencies of all the alleles of each one of the five loci, the following parameters have been calculated:

- "A priori" Probability of Exclusion for every locus (P_{locus}). This value gives the efficiency of the locus studied in paternity exclusion.

- Accumulated Chance Exclusion, gives the efficiency of the five loci in paternity exclusion.

$$P_{accum} = 1 - (1 - P_{D1S80}) \times (1 - P_{YNZ22}) \times (1 - P_{3'ApoB}) \times (1 - P_{ACTBP2}) \times (1 - P_{HLA\ DQ\alpha})$$

- The Essen Möller value is the biostatistical efficiency of the locus.

$$E.M. = 10 + \log Y/X$$

- Accumulated Essen Möller value is the biostatistical efficiency of the loci:

$$E.M._{accum} = \frac{EM_{D1S80} \times EM_{YNZ22} \times EM_{3'ApoB} \times EM_{HLA\ DQ\alpha} \times EM_{ACTBP2}}{10^4}$$

RESULTS AND DISCUSSION

Table 1 shows the probability of exclusion and the Essen Möller value obtained for the five loci independently and as a whole.

Locus	P.excl.	E.M.value
ACTBP2	0.8808	9.0108
YNZ22	0.6977	9.3561
D1S80	0.6367	9.3323
HLA DQα	0.6072	9.5361
3'ApoB	0.5886	9.4717
Five loci	0.9978	7.1064

Table 1. Probability of exclusion and biostatistical efficiency of the five loci studied by PCR in the population of Catalonia.

DISCUSSION

In our population the best performance of all genetic markers is obtained by the ACTBP2. The results obtained by the other four PCR polymorphism studied are only improved by those of the loci HLA A and B. Table 2 shows the relation between these five genetic markers and some of the main classical systems.

Locus	P.excl.
1 ACTBP2	0.88
2 HLA B	0.85
3 HLA A	0.78
4 YNZ22	0.69
5 D1S80	0.63
6 HLA DQα	0.60
7 3'ApoB	0.58
8 HLA C	0.35
9 Pi	0.34
10 PGM 1	0.33

Table 2. Probability of exclusion "a priori" of the first ten genetic markers in the population of Catalonia.

Table 3 shows the comparison between the different groups of polymorphisms traditionally used and the PCR group.

Polimorphism group	Chance exclusion
PCR	0.99
HLA ABC	0.98
RBC	0.73
EE	0.61
PP	0.58

Table 3. Probability of exclusion of different polimorphism groups used in paternity testing.

CONCLUSIONS

In the population of Catalonia the locus ACTBP2 shows the highest power of exclusion of all the markers we use in paternity testing.

The exclusion probability of the PCR group is 0.9978, and offers the best statistical values compared with the other genetic marker method groups used in paternity testing in the population of Catalonia.

The accumulated value of chance exclusion obtained by using PCR, HLA ABC, RBC, EE and PP is over 0.9999

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