

FRENCH POPULATION DATA OF 6 AMPFL'S

PASCAL O. , LEVAYER T. , AUBERT D. , PENEAU A. , MARKEY P. , MOISAN J.P.

Laboratory of Molecular Genetics . University Hospital . NANTES FRANCE

Introduction :

The Polymerase Chain Reaction (PCR) is a powerful tool for DNA typing. This technique is useful because of its sensitivity, speed and ability to type few or partially degraded DNA molecules. In order to apply PCR routinely, population data is needed to estimate genotype frequencies. We present here the results obtained for French caucasian people in seven systems. In addition, we point out some applications.

Materials and methods :

DNA was extracted from blood samples and amplified in a Cetus thermocycler as previously described : D1S80 (1), ApoB (2), YNZ 22 (3), COL 2A1 (4), TC11 (5), von Willebrand Factor (6), HLADQ α (7). PCR products were analyzed in horizontal polyacrylamide gel (1) (D1S80, ApoB, YNZ22, COL2A1, TC11), in vertical polyacrylamide gel (von Willebrand Factor) or using non radioactive detection (HLADQ α) (7). 110 unrelated caucasian people were analyzed for the population data.

Results :

HLADQ α :

Alleles	Frequency
1.1	16.4 %
1.2	17.3 %
1.3	8.0 %
2	20.8 %
3	13.2 %
4	24.3 %

TC11:

Allele	Frequency:
1 (lower)	23.8 %
2	15.2 %
3	12.9 %
4	17.5 %
5	30.2 %
6 (upper)	0.4 %

D1S80 :

Allele	Frequency
16	0.0 %
17	0.4 %
18	26.3 %
19	1.5 %
20	1.9 %
21	2.2 %
22	6.3 %
23	1.1 %
24	30.7 %
25	6.3 %
26	2.6 %
27	1.5 %
28	5.2 %
29	2.9 %
30	1.9 %
31	6.7 %
32	1.9 %
>32	0.0 %

YNZ22 :

Allele	Frequency
1 (lower)	6.6 %
2	24.1 %
3	9.3 %
4	25.9 %
5	4.8 %
6	3.7 %
7	0.0 %
8	5.2 %
9	5.9 %
10	6.3 %
11	4.8 %
12	2.6 %
13 (upper)	0.8 %

Von Willebrand Factor :

Allele	Frequency
1 (138 bp)	9.7 %
2 (142 bp)	10.5 %
3 (146 bp)	20.2 %
4 (150 bp)	31.9 %
5 (154 bp)	18.9 %
6 (158 bp)	7.9 %
7 (162 bp)	0.9 %

ApoB :

Allele	Frequency
1 (lower)	6.8 %
2	0.4 %
3	6.1 %
4	27.7 %
5	37.5 %
6	3.8 %
7	2.3 %
8	0.7 %
9	4.5 %
10	6.1 %
11	3.4 %
12 (upper)	0.7 %

Discussion :

Southern blotting is, for DNA typing the routine technology. However southern blotting could be unsuitable in case of a low amount of DNA or partially degraded DNA. In a such situation the Polymerase Chain Reaction (PCR) is the only technique possible for typing DNA.

Cigarette butts are routinely analyzed in our laboratory. A minimal amount of DNA (i.e. 1 ng) is obtained in about 50% of samples, making possible the DNA amplification in, at least, one system. Then genotype frequency will depend on the number of system used.

Formalin treated or paraffined samples are not suitable for southern blotting. We succeed in DNA amplification for such samples. The extraction procedure is the major part of the process : a combination of classic extraction (proteinase K, phenol-chloroform) and further purification steps (Chelex[®], Centricon[®], dialysis) gives the best results.

DNA amplification is an attractive technique which seems simple, quick and inexpensive. However several problems are critical : unequal allele amplification in some systems, presence of inhibitors, allele marker standardisation, contaminations and low discrimination power.

In our laboratory a few cases have been resolved by DNA amplification but technique credibility has not been estimated since no trial has been judged with results obtained by PCR. In our opinion, today, Southern blotting must be the reference procedure for Forensic DNA typing. However the Polymerase Chain Reaction could be used in case of failure of the other techniques.

References :

- 1 Budowle B., Chakraborty R., Giusti A., Eisenberg A., Allen R.
Am. J. Hum. Genet. (1991) **48** : 137 - 144
Analysis of the VNTR locus D1S80 by the PCR followed by high resolution PAGE.
- 2 Boerwinkle E., Weijun X., Fourest E., Lawrence C.
Proc. Natl. Acad. Sci. USA (1989) **86** : 212-216
Rapid typing of tandemly repeated hypervariable loci by polymerase chain reaction: application to the apolipoprotein B 3' hypervariable region.
- 3 Rand S., Puers C., Skowasch K., Wiegand P., Bodowle B., Brinkmann B.
Int. J. Leg. Med. (1992) **104** : 329 - 333
Population genetics and forensic efficiency data to 4 AMPFLP'S.
- 4 Wu S., Seino S., Bell G.
Nucleic Acids Res. (1990) **18** : 3102
Human collagen, type II, alpha 21, (COL2A1) gene : VNTR polymorphism detected by gene amplification.
- 5 Edwards A., Civitello A., Hammond H., Caskey T.
Am. J. Hum. Genet. (1991) **49** : 746 - 756
DNA typing and genetic mapping with trimeric and tetrameric tandem repeats.
- 6 Kimpton C., Walton A., Gill P.
Hum. Mol. Genet. (1992) **1** : 287
A further tetranucleotide repeat polymorphism in the vWF gene.
- 7 Amplitype[™] Cetus Corporation
Catalog N°808-0056