

# Chromosome polymorphisms in paternity investigations

V.M. Almeida, A. Amorim

Instituto de Antropologia, Universidade do Porto, Portugal

## INTRODUCTION

The use of chromosome polymorphisms in paternity expertises has been rare, when compared with current classical antigen or biochemical analysis or even with HLA phenotyping. In this work we demonstrate that polymorphism of heterochromatic regions of chromosomes 1, 9 and 16, evidenced by simple routine cytogenetic techniques, may become very helpful, at least in "difficult" cases, such as those where a man is not excluded using current battery of tests but the corresponding value of W is low.

## MATERIAL AND METHODS

The polymorphism of heterochromatic regions of chromosomes 1, 9 and 16 was studied in a sample (n=302) of unrelated individuals from Porto district (Portugal).

Metaphase chromosomes were obtained by a 72 hours lymphocyte cell culture (Moorehead et al.,1960). Slides were stained by the C-banding technique of Sumner (1972). At least eight, and up to fifteen metaphases were observed per individual.

## RESULTS

The common polymorphisms of heterochromatic regions of chromosomes 1, 9 and 16 are depicted in Fig.1.

In Table 1 we present the corresponding frequencies in Porto district as well as the *a priori* probability of exclusion (PE) and the probability of paternity among fathers (WF).

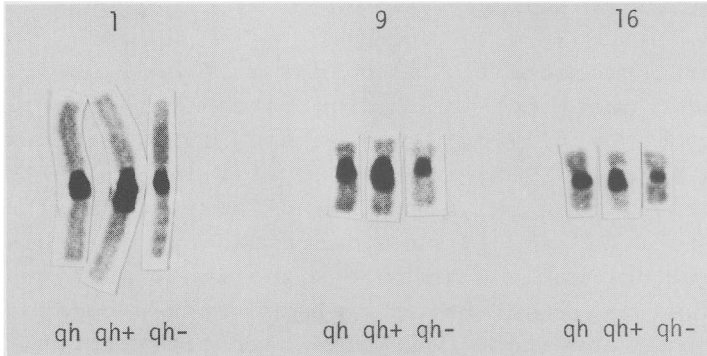


Fig 1. Polymorphism of heterochromatic regions of chromosomes 1, 9 and 16 observed in our sample.

Table 1. Frequencies of chromosome polymorphisms, *a priori* probability of exclusion (PE) and mean probability of paternity among fathers (WF).

chromosome		freq.(%)	PE(%)	WF(%)
1	1qh+	3.3	7.7	54.1
	1qh-	4.8		
	1qh	91.9		
9	9qh+	4.3	6.2	53.3
	9qh-	2.2		
	9qh	93.5		
16	16qh+	3.3	3.6	52.0
	16qh-	0.5		
	16qh	96.2		
TOTAL			16.6	59.3

## DISCUSSION

The study of chromosome polymorphisms is highly useful in many fields of human genetics, including paternity investigations: (Gürtler and Niebuhr, 1986; Olson et al., 1986; Balícek et al., 1978; Shiono et al., 1985; Kamenev et al., 1986). Although not very informative, the polymorphisms of heterochromatic regions of chromosomes 1, 9 and 16 here described can be useful, at least as ancillary markers in some situations, such as:

- (a) the accused man, not being excluded by the current battery of tests of the laboratory has a low paternity probability and,
- (b) deceased persons are involved, but their karyotypes are available, namely resulting from previous clinical studies.

REFERENCES

- Balícek P, Zizka J, Skalská H (1978) Variability and familial transmission of constitutive heterochromatin of human chromosomes evaluated by the method of linear measurement. *Hum Genet* 42: 257-265
- Gürtler H, Niebuhr E (1986) Chromosome polymorphisms in legal paternity cases. In: Brinkman B, Henningsen K (eds) *Advances in Forensic Haemogenetics*, Spriger-Verlag, Berlin, p 201-211
- Kamenev LE, Andre AC, Frederic JL (1986) Efficiency in disputed paternity cases of a new category of markers: chromosome variants. In: Brinkman B, Henningsen K (eds) *Advances in Forensic Haemogenetics*, Spriger-Verlag, Berlin, p 411-414
- Moorehead PS, Nowell PC, Mellman WJ, Battips DM, Hungerford DA (1960) Chromosome preparations of leucocytes cultured from human peripheral blood. *Exp Cell Res* 20:613
- Olson SB, Magenis RE, Lovrien EW (1986) Human chromosome variation: the discriminatory power of Q-band heteromorphism (variant) analysis in distinguishing between individuals, with specific application to cases of questionable paternity. *Am J Hum Genet* 38: 235-252
- Shiono H, Azumi J, Sakamoto Y (1985) Chromosome heteromorphisms and paternity testing. *Am J For Med Path* 6: 199-203
- Sumner AT (1972) A simple technique for demonstrating centromeric heterochromatin. *Exp Cell Res* 75: 304-306