

International Congress Series 1239 (2003) 861-864

Incestuous offspring detection inference by VNTR homozygosity increment

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Abstract

Sexual relations between parent and offspring and between sister and brother are defined as incest. People involved are legally prohibited from marrying each other because of their close relationship. Usually, when children are involved, violence and sexual abuse are committed. Recent studies have indicated that incest, although rarely discussed, is an increasing problem in modern society. Prohibition of incest as a form of social behaviour may rely on a genetic basis. It is known that inbreeding fosters the concentration of undesirable recessive genes. The only unbiased approach for incestuous offspring identification is DNA typing. In some cases, incest detection may arise during a regular paternity test. The aim of this contribution is to present the comparative analysis of incest and non-incest cases. This contribution may facilitate the interpretation of cases in which the incest victim is unavailable.

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Keywords: Incest; VNTR; Paternity testing

1. Introduction

Almost universally, sexual relations between individuals closely related is defined as "incest". Sexual molestation by family members is a recurring form of child abuse. Inbreeding fosters the concentration of undesirable recessive traits and reduces the heterozygosity levels in polymorphic genomic sequences. In the forensic field, the statutory crime of incest consists of either cohabitation or sexual intercourse between

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closely related persons, such as between father and child or between siblings. As a special case of paternity testing, the unique, unbiased approach for incestuous offspring identification is DNA typing. In addition, this technique may also provide information that can allow the inference of an incestuous origin for an offspring even in the absence of the biological mother. The aim of this contribution is to present the comparative analysis of incest and non-incest cases, involving fathers–daughters, investigated by means of diverse VNTR platforms.

2. Materials and methods

Incestuous cases: 33 incestuous cases were analysed. All of them were trios, mother (M), child (C) and alleged father (AF), where the AF is also the father of M. This condition was checked after genetic typing. Eleven cases were analysed with both mini and microsatellites. Twenty-three cases were analysed only by means of microsatellites included in commercial kits.

Control cases: More than 300 paternity tests, including M, C and AF were used as "control cases", in which no relationship between M and AF was the condition for use.

2.1. Loci investigated

Minisatellites: probes YNH-24, MS-1, LH-1, PH-30, TBQ-7, EFD-52 and CEB-42, analyzed by Southern blot.

Microsatellites typed using manual techniques: amplicons of the followings STRs: HUMTHO-1, HUMFABP, TPOX, CSF1PO, VWA, F13AO1, FES/FPS, D16S539, D7S820, D13S317 and D6S366 were resolved by electrophoresis in denaturing gels followed by silver staining.

Automated STR typing: microsatellites included in the commercial kits: Profiler Plus and Cofiler (Applied Biosystems) and Powerplex 16 (Promega) were analyzed using the ABI Prism 310 sequencer.

Homozygosity levels and heterozygous patterns identical to the mother were counted in the offspring.

3. Results and discussion

The results of incestuous cases are summarized in Figs. 1 and 2. To compare the homozygosity levels in control and incestuous families, cases were divided into those that were investigated by means of mini and microsatellites, and those that were analyzed only by microsatellites. Higher levels of homozygosity are detected in incestuous cases compared with control cases. More than 60% of cases detected homozygosity levels higher than 40%, and in contrast, in control cases more than 80% show homozygosity levels lower than 30%. High homozygosity percentages in the offspring could modify

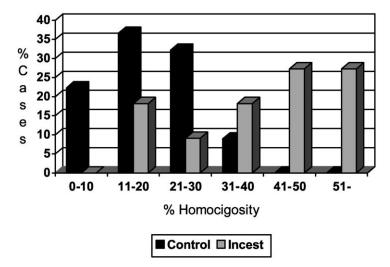


Fig. 1. Levels of homozygosity in control and incest cases analyzed by means of micro and minisatellites.

the total paternity index in those situations in which the mother is not available for analysis [1].

The frequency distribution of homozygous markers is completely skewed when compared with non-incestuous offspring. Although some overlap may be detected when the incestuous/non-incestuous distribution of homozygotes are compared, if the homo-

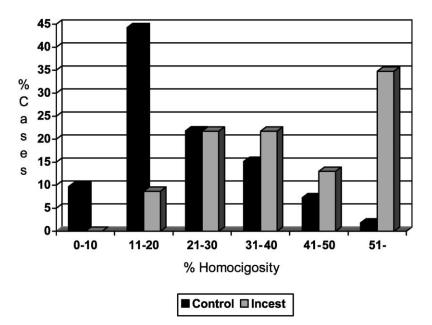


Fig. 2. Levels of homozygosity in control and incest cases analyzed by means of microsatellites.

Table 1

Comparison of homozygosity levels and profiles heterozygous and equal to the mother in the descendent (control and incestuous cases)

	Mini and microsatellites		Microsatellites	
	Control	Incestuous	Control	Incestuous
% Homozygous	17.38	42.11	21.25	42.17
% Heterozygous and equal	12.61	32.03	16.91	25.67

zygosity percentage is over 40%, the offspring can be confidently considered as incestuous.

Table 1 summarizes the average percentage of homozygosity as well as heterozygous patterns identical to the mother found in the offspring.

In our laboratory, since 1992, over 940 paternity cases were investigated and only 33 were demonstrated as incest cases (about 3, 5%). This figure does not reflect the real situation: many incest situations are not declared.

In conclusion, in paternity cases in which father and mother are directly related, the homozygosity levels are higher than those in which father and mother are unrelated. The observation of homozygosity levels higher than 40% could be considered as a signal of incest, in those cases in which the mother is not available and the incest is not declared.

Reference

[1] C. Brenner, A note on paternity computation in cases lacking a mother, Transfusion 33 (1993) 51-54.