



The number of STR markers necessary to resolve relationships in deficiency paternity cases

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Abstract. The inference about the biological relationship between pairs of individuals using genetic markers plays a central role in many areas of human genetics. We investigated the number of markers (M) that are necessary to assign a given proportion of pairs (50%, 60%, 70%, 80%, 90%, 95%, and 99%) to their correct relationship at three predefined probability levels (99%, 99.9%, and 99.99%) against several alternative hypotheses. The following relationships were considered: (1) full sibs (FS), (2) second degree (2D, including half-sibs, grandparent–grandchild and avuncular pairs), (3) first cousins (FC), (4) unrelated individuals (UR). © 2003 Published by Elsevier B.V.

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1. Introduction

Deficiency paternity cases are not unusual in the forensic practice. Typically, these situations arise when the alleged parent of a claimant is not available (in most cases this person is deceased), and one of his or her undisputed relatives acts as a defendant, i.e. s(he) denies claimant's contentions. Most of these cases reduce, at least at first examination, to determine the likelihood ratio of two alternative hypotheses about the relationship between a single pair of individuals. We have encountered instances in which the contrasting propositions about the two contenders were as follows: half-sibs vs. unrelated; avuncular pair vs. unrelated; full sibs vs. half-sibs; first cousins vs. unrelated; full sibs vs. unrelated. Here, we addressed the question of the number of autosomal DNA markers needed to resolve common relationships at specified probability levels by computer simulations.

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3. Results

Table 1 shows the number of markers necessary to assign a given percentage of pairs (second row) to each of six possible relationships (first column) at the specified level of probability (second column).

4. Discussion

Full sibs (FS) are discriminated from nonrelatives (UR) and first cousins (FC) with a reasonable number of markers; for instance, 90% of FS pairs are discriminated from UR pairs with probability of 99.99% using 18 markers (which are easily found in commercial kits); the same number of markers allows discriminating more than 50% of FS from FC at the same probability level. Discrimination between full and half sibs (FS/HS) is more problematic, in accordance with published data [3,4]; 18 markers can solve more than 50% of cases but only with a probability of 99%. First cousins (FC) are poorly discriminated from second degree (2D) and nonrelatives (UR), even in the hypothesis of the availability of 50 validated markers. Future extensions of this work will include the use of X- or Y-chromosome markers, which can allow solving particular cases of disputed relationships (for instance, two males that share the same Y-haplotype have a very low probability of being non relatives [5,6], whereas X-chromosome markers provide paternity indexes higher than similar autosomal markers in the case of female pairs).

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