International Congress Series 1239 (2003) 831–833

Results of the 2001 paternity testing workshop of the English speaking working group

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Abstract

We present the results of the 2001 paternity testing workshop of the English speaking working group of the International Society for Forensic Genetics. In total, 36 laboratories participated. The scenario was an alleged father and two children who were known to be children of the same non-investigated mother. All laboratories drew the correct conclusion. The laboratories used a total of 58 PCR-based systems, 14 conventional systems and 10 RFLP-based systems. The workshop also included a paper challenge. The results here revealed that occurrences of rare alleles, mutations and possible silent alleles were treated very differently among the participating laboratories.

Keywords: Paternity testing; DNA profiling; Collaborative exercise

1. Introduction

Since 1991, the English Speaking Working Group (ESWG) of the International Society for Forensic Genetics (ISFG) has, once a year, offered an exercise involving genetic analysis in a paternity case [1–3]. The collated results of the exercises include typing results and information about laboratory routines, systems and kits used for paternity testing as well as information about statistical calculations.

Since 1998, laboratories performing immigration cases have been invited to treat the case also as they would do in an immigration case. Since the year 2000, the laboratories have been invited to calculate a paper challenge in addition to the paternity testing.

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0531-5131/03
PII: S0531-5131(02)00299-6

2. Materials and methods

Blood samples from two children and an alleged father were sent to the laboratories together with the information needed to treat the case as a paternity case as well as an immigration case. A questionnaire concerning the techniques and routines in the laboratories was distributed. A total of 36 laboratories participated in the workshop.

3. Results

Results from a total of 58 PCR-based systems, 14 conventional systems and 10 RFLP-based systems were submitted. Of these, results from 36 PCR-based systems, two conventional systems and nine RFLP-based systems were submitted by more than one laboratory.

The results showed that most laboratories used the same nomenclature. Among the PCR-based systems, results with inconsistent nomenclature were submitted in two systems.

As the laboratories used a large number of different systems and as they performed statistical calculations based on their own frequency databases, comparison of the calculated PI-values was not possible. For that purpose, the exercise included a paper challenge. The paper challenge was designed to include rare events such as rare alleles, mutations and possible silent alleles. Furthermore, database information on the alleles was given as numbers of observations of the alleles in the database and all calculations were left to the laboratories. A total of 22 laboratories reported PI-values (or other statistical values) for each system in the paper challenge. A total of 21 different cumulative PI-values were obtained. Most discrepancies were due to different calculations in the systems with the rare events. Depending on the exact circumstances, 64–82% of the laboratories did not consider silent alleles. In one system, a maternal inconsistency was present. A total of 13% of the laboratories did not calculate a paternity index for that system. Among the rest, a total of nine different formulas was used for calculations. In one system, ‘Man’ and ‘Child’ shared an allele that was not present in the database. For calculations, 55% of the laboratories used a fixed minimum frequency while 32% used a frequency of 1/N. A total of 9% of the laboratories used a frequency based on the sum of observations of the allele in the present case and 5% did not calculate a paternity index. A total of 62% of the laboratories concluded that the results were in favour of paternity with a paternity index $> 10\,000$. A total of 4% of the laboratories concluded that results were in favour of paternity with a paternity index>1000 while 35% of the laboratories recommended further testing.

4. Discussion

While methods and nomenclature have reached a high degree of standardisation, the results of the paper challenge showed that statistical calculations of paternity indices varied when seldom events such as rare alleles, mutations and possible silent alleles were present.

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