



# Results of the 2003 paternity testing workshop of the English Speaking Working Group of the International Society of Forensic Genetics

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**Abstract.** We present the results of the 2003 paternity testing workshop of the English Speaking Working Group (ESWG) of the International Society of Forensic Genetics (ISFG). The scenario was an alleged father, one child and the mother. All 51 participating laboratories drew the correct conclusion. The laboratories used a total of 30 autosomal and 17 Y-chromosomal PCR-based STR systems and 7 RFLP-based VNTR systems. The percentage of typing errors was 0.12% for the autosomal systems and 2% for the Y-chromosomal systems. The results from a paper challenge showed that occurrences of rare events such as mutations and possible silent alleles were treated differently among the participating laboratories. Participating laboratories reported mutational events observed in STR systems. Considerable differences between mutation rates in STR loci were reported. There were marked differences between paternal and maternal mutation rates as well as between single-step and multiple-step mutation rates. © 2003 Elsevier B.V. All rights reserved.

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## 1. Introduction

Since 1991, The English Speaking Working Group (ESWG) of the International Society of Forensic Genetics (ISFG) has offered an annual exercise involving genetic analysis of a paternity case [1–4]. 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 the year 2000, the laboratories have been invited to calculate a paper challenge in addition to the paternity testing.

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## **2. Materials and methods**

Blood samples from a mother, a child and an alleged father were sent to the laboratories together with the information needed to treat the case as a paternity case. A questionnaire concerning the techniques, routines and genetic systems used by the laboratories was distributed. Fifty-one laboratories submitted their results.

## **3. Results**

All laboratories concluded that their results were in favour of paternity. Results from a total of 30 autosomal PCR-based systems, 17 Y-chromosomal PCR-based systems and 7 RFLP-based systems were submitted. Of these, results from 26 autosomal PCR-based systems, 12 Y-chromosomal PCR-based systems and 6 RFLP-based systems were submitted by more than one laboratory.

The results showed that most laboratories used the same nomenclature. Among the somatic PCR-based systems, no inconsistencies were due to different nomenclature. Among the Y-chromosomal PCR-based systems, results with inconsistent nomenclature were submitted in two systems (DYS389I and *DYS389II*).

The typing error rate of the submitted results was 0.12% for the autosomal systems and 2% for the Y-chromosomal systems. All laboratories used one or more commercially available PCR-based STR-typing kits for typing of autosomal systems. In contrast, less than 50% of the labs used commercially available kits for Y-STR typing.

The exercise included a paper challenge with calculation of paternity indices of rare events such as mutations and possible silent alleles and with Y-chromosomal haplotypes. Information was given on the number of observations of the relevant alleles in the database and all calculations were left to the laboratories. Thirty-one laboratories reported PI-values (or other statistical values) for each autosomal system and 23 laboratories included PI-values (or other statistical values) for the Y-chromosomal haplotype. Twenty-six different cumulative PI-values were obtained. Most discrepancies were due to different calculations of the systems with inconsistencies or possible silent alleles. Most laboratories (92%) concluded in the paper challenge that results were inconclusive and recommended further testing, 5% concluded that results were in favour of paternity and 3% that results were against paternity.

Laboratories were asked to report mutation events of STR systems. The overall mutation rate of the autosomal STR systems was 0.114% (range <0.001–0.780%). The overall paternal mutation rate was 0.153% and the overall maternal mutation rate was 0.043%. The overall mutation rate for single-step mutations was 0.109% and the overall mutation rate for multiple-step mutations was 0.005%.

## **4. Discussion**

The use of commercially available STR kits seems to have facilitated a high level of standardisation of methods and nomenclature among the participating laboratories in the paternity testing workshop. The typing error rate of the submitted results was 0.12% for the autosomal systems and 2% for the Y-chromosomal systems. All participating laboratories used one or more commercially available kits for autosomal STR systems

but less than half of the reporting laboratories used commercially available kits for Y-STR typing.

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. The results also showed that there is no consensus on statistical calculations of Y-chromosomal haplotypes.

## References

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