

## Genetic inconsistencies in paternity cases

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**Abstract.** Knowledge about mutation rates and mutational process of short tandem repeats (STRs), microsatellite loci used in paternity testing and forensic analysis, is crucial for the correct interpretation of resulting genetic profiles. Therefore, this study was carried out in 629 paternity cases, from 2000 to 2003, in which more than 2000 individuals were studied and genetic inconsistencies discovered during biological relationship testing. © 2003 Elsevier B.V. All rights reserved.

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### 1. Introduction

Short tandem repeats (STRs) polymorphisms have become a powerful tool for human identification, and actually are the genetics markers more useful. However, as the STR markers are more mutable than the classical serological or molecular markers, parentage-testing laboratories are observing sporadic exclusion caused by mutations during maternal or paternal meiosis events.

Genetic inconsistencies discovered during biological relationship testing can add complexity to the analysis and resolution of a case [4–6].

### 2. Materials and methods

This study was carried out in 629 paternity cases, from 2000 to 2003 in which more than 2000 individuals were studied.

Genomic DNA was isolated from blood samples with salting out Miller's method [15]. The samples were amplified at the loci FGA, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D18S51 and D21S11 using the AmpFISTR® Profiler Plus Kit (PE Biosystems, Foster City, CA) and at the loci CF1PO, TPOX, TH01, D3S1358, D7S820, and D16S539 using the AmpFISTR® Cofiler™ kit (PE Biosystems). The same loci and the D2S1338 and D19S433 were amplified with the AmpFISTR® Identifier™ Kit (PE Biosystems). Samples were analyzed using the ABI PRISM® 310 Genetic Analyzer (PE Biosystems), according to the manufacturer's recommendations using as separation medium Performance

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Table 1  
The mutation rates at the individual STR loci

Locus	No. of meiosis		No. of mutation		Mutation rate		Type of mutation
	Male	Female	Male	Female	Male	Female	
vWA	562	309	1	–	0.18%	–	Addition
CSF1P0	562	309	1	–	0.18%	–	Deletion
D7S820	562	309	1	–	0.18%	–	Addition
D21S11	562	309	2	–	0.35%	–	Addition (2)
D8S1179	562	309	1	–	0.18%	–	Addition
FGA	562	309	1	–	0.18%	–	Deletion
D13S317	562	309	1	–	0.18%	–	Null allele
D2S1338	562	309	1	–	0.18%	–	Deletion
D3S1358	562	309	1	–	0.18%	–	Null allele

Optimized Polymer (POP) 4TM (PE Biosystems). The data was acquired by 1.0.2 software ABI PRISM® 310 Collection and analyzed by GeneScan® Analysis software 3.1 and Genotyper® 2.5 according to the manufacturer's recommendations [1–3,13,14].

### 3. Results

From more than 600 studies, only 1.6% showed inconsistent results for one of the locus investigated. These inconsistencies were present at vWA, CSF1P0, D7S820, D8S1179, FGA, D13S317, D2S1338 and D3S1358 loci. The mutations observed were the following ones:

1. Most of them were either gains or losses of complete repeats.
2. The number of additions was significantly major than the number of deletions.
3. We assume the presence of null alleles at D3S1358 and D13S317 in two paternity cases where the alleged father had a homozygous genotype.

The mutation rates at the individual STR loci are given in Table 1.

In these cases, we made the mathematical and statistical calculations of Paternity Probability with and without the presence of the mutation. In all of the cases, the percentage of Paternity Probability [10–12], including the inconsistency, was greater than 99,99% [7–9].

### 4. Conclusion

For this number of investigated markers, our exclusion criteria is based on the presence of more than three inconsistencies in the paternity investigation.

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