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STR mutations in paternity investigations: a study of 1-year consecutive cases

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Abstract

A study of 1-year consecutive cases for STR mutations has been performed during 2000 with SGM Plus and PowerPlex 1.2 or PowerPlex 16. In paternity investigation cases, 18 STR mutations have been detected in several loci. Paternal and maternal mutation cases and a case with two paternal inconsistencies in D7S820 and CSF1PO are also presented. Maternal and paternal meioses were studied and the average mutation rates concerning several types of mutations were determined. During this 1-year study, multi-banded alleles and vWA genotype differences using different multiplex systems were also detected. The final probability paternity value (after inclusion of mutations) was always >99.99%, allowing to conclude confidently the presence of a mutation.
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1. Introduction

Genetic inconsistencies detected during paternity investigation studies can add complexity to the analysis and resolution of these investigations. Since STR systems are nowadays the most commonly used systems in paternity cases, the chance of detecting mutations increases as STRs have mutation rates higher than conventional genetic markers.

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Since 1998, only STR loci have been used in our laboratory to study paternity investigation cases, with an average of 500 cases per year. During 2000, paternity investigation cases were studied with SGM Plus and PowerPlex 1.2, typing a total of 15 STR loci. At the end of the year, PowerPlex 16 was introduced replacing PowerPlex 1.2, giving a total of 17 STR loci—the 13 Codis loci plus D2S1338, D19S433, Penta E and Penta D.

A study of 1-year consecutive cases for STR mutations in paternity investigations has been conducted with these three multiplex systems. The number of cases of mutation encountered accounts for 3% of the total paternity investigation cases performed during 2000 and several different types of mutations were detected. Maternal and paternal meioses were studied in the 17 STR loci and the average mutation rates were also determined. With the increase of STR loci number and due to the high mutation rates detected in STR loci, we can expect to find paternity investigation cases with two or even three genetic incompatibilities that are non-exclusion cases.

2. Materials and methods

Portuguese Caucasian individuals mainly from South Portugal were analysed from 511 paternity investigation cases. Blood samples were collected in Ultra Stain Card, extracted with Chelex and purified with Wizard DNA Clean-Up System. Amplifications were performed with a Perkin Elmer GeneAmp PCR System 9600 and electrophoresis was carried out in an ABI Prism 377 DNA sequencer, according to the manufacturer's instructions for SGM Plus and PowerPlex 1.2 or PowerPlex 16.

3. Results

Using SGM Plus and PowerPlex 1.2/16 multiplex systems, 18 mutations have been detected in 13 STR loci. Maternal and paternal meioses studied, genetic inconsistencies

Table 1
Paternal and maternal genetic inconsistencies cases detected in several STR loci

Loci	*W* without mutation	A. Father	Mother	Child	*W* with mutation
D3S1338	99.99997	18–*18*	14–16	16–*17*	99.995
vWA	99.99995	18–*19*	16–19	19–*20*	99.993
D8S117	99.99999990	14–*14*	12–14	12–*15*	99.99998
	99.99999995	***12***–13	14–15	***10***–14[a]	99.9998
D13S317	99.999999997	12–*12*	12–13	13–*13*[b]	99.9999996
D2S1338	99.9999999997	*23*–24	20–23	20–*22*	99.99999994
	99.999995	18–23	20–*20*	18–*18*[c]	Maternal mutation
D16S539	99.9999999995	*11*–*13*	9–13	9–*12*	99.99999992
D19S433	99.999996	13–15	13–*17*	15–*16*	Maternal mutation

[a] Possible two-step mutation.
[b] One-step mutation or null allele.
[c] Possible null allele.

Table 2
Number of meiosis studied and mutations and mutation rates detected in 17 STR loci in a 1-year consecutive cases study

Loci	Meiosis number		Mutation number		Mutation rate ($\times 10^{-3}$)		
	Maternal	Paternal	Maternal	Paternal	Maternal	Paternal	Total
D3S1358	378	311	0	1	–	3.2	1.5
vWA	378	311	0	2[a]	–	6.4	2.9
D16S359	378	311	0	2	–	6.4	3.0
D2S1338	350	293	1[b]	1	2.9	3.4	3.7
D18S51	376	311	0	1	–	3.2	1.5
D8S1179	378	311	0	3[c]	–	9.7	4.4
D21S11	378	311	0	1	–	3.2	1.5
D19S433	350	293	1	0	2.9	–	1.6
D13S317	378	311	0	1	–	3.2	1.5
D7S820	372	311	0	1	–	3.2	1.5
D5S818	372	311	0	0	–	–	–
CSF1PO	368	307	0	1	–	3.3	1.5
Penta E	80	70	0	1	–	14.2	6.7
FGA	378	311	0	1	–	3.2	1.5
Penta D	80	70	0	0	–	–	–
TH01	378	311	0	0	–	–	–
TPOX	368	309	0	0	–	–	–
Total	5740	4763	2	16	Average 0.348	Average 3.36	Average 1.71

[a] One mutation can either be maternal or paternal.
[b] Possible null allele.
[c] One case with a two-step mutation.

detected and maternal, paternal and total mutation rates observed in 17 STR loci are outlined in Table 1. Example cases with one paternal or maternal genetic inconsistency and respective *W* values are presented in Table 2.

In this study, a paternity case with two paternal genetic inconsistencies in D7S820 and CSF1PO was also detected. Twenty-two different STR loci were then used—13 Codis loci, D2S1338, D19S433, Penta E, Penta D, D19S253, D12S391, D1S1656, FES/FPS and F13A1. A one-step mutation difference was assumed in both loci (Table 3).

We have also detected three cases with a multi-banded pattern [1]—two cases with a multi-banded pattern of maternal origin in TPOX (genotype 8-10-11) and one case with a multi-banded pattern of paternal origin in D18S51 (genotype 13-14-16). A vWA genotype difference in one case was also detected when PowerPlex 1.2 and SGM Plus were used to

Table 3
A paternity case with two paternal genetic inconsistencies in D7S820 and CSF1PO

W without mutations (99.999999994%)	A. Father (26Y)	Mother (28Y)	Child	*W* including mutations (99.9998%)
D7S820	10–***13***	9–12	12–***14***	
CSF1PO	9–***13***	12–13	12–***12***	

amplify the same samples—a homozygous 16–16 detected with SGM Plus produced a heterozygous 16–17 with PowerPlex1.2. This is due to the different primer sets used in each system [2].

4. Conclusions

In all mutation cases encountered in this investigation, there is very strong evidence in favour of paternity and maternity. Mutations detected in this study present the following characteristics:

- The majority of mutations can be assumed to involve the addition or loss of a single repeat unit and the overall STR mutation rate is 1.71×10^{-3} considering 18 mutations when studying 10,503 total meioses [3].
- The average paternity mutation rate was 3.36×10^{-3}, 10 times higher than the maternal one (3.48×10^{-4}).
- One paternal genetic inconsistency involved the deletion of two repeat units in D8S1179, with an average two-step paternal mutation rate of 2×10^{-4}. With paternal mutations, one-step mutation occurred 15 times more frequently than two-step mutations.
- A null allele was assumed to have occurred in the D2S1338 locus in a maternal incompatibility due to non-matching homozygous patterns between the mother and child.
- Genetic inconsistencies in TH01, TPOX, D5S818 and Penta D were not detected during this study.
- The final probability of paternity in these paternity investigations (after inclusion of mutation in statistical calculations) was always >99.99%, allowing to conclude the presence of a mutation.

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