

An evaluation of rapidly mutating Y-STR multi-allelic markers



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ARTICLE INFO

Article history:

Received 27 August 2015
Received in revised form 15 September 2015
Accepted 7 October 2015
Available online 20 October 2015

Keywords:

Y-STR
Forensic
E-type
C-Type
Multi-allelic

ABSTRACT

Multi-allelic Y chromosome short tandem repeats (Y-STRs) have been avoided in forensic applications due to high level of complexity involved in the interpretation of these markers. Because of such reason Y-STR multi-allelic markers have less representation in literature compared to Y-STR single-allelic markers. However these markers were proven to be highly polymorphic previously such as DYS464. In the recent discovery of the 13 rapidly mutating Y-STRs there were four multi-allelic markers including DYF387S1, DYF399S1, DYF403S1 and DYF404S1. In all subsequent studies these markers were always showing the highest diversity across the 13 RM Y-STRs. In this study these markers were investigated individually in 361 male individuals from United Arab Emirates. Each marker was analyzed using previously published primers sets. The potential forensic application value of these markers was evaluated using both conservative (C-type) and expanded (E-type) approaches.

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1. Introduction

The high power of discrimination resulting from typing multi-allelic Y-STR marker is only advantageous when analysing single source DNA, where as these markers cannot be interpreted easily when more than one male are contributing to the DNA sample [1]. Another factor which can also complicate the analysis is the imbalanced peak height in such markers. Previously an analysis of DYS464 marker was conducted to estimate the efficiency of using both conservative (C-Type) and expanded (E-Type) genotyping approaches [2]. In this study, the analysis of recently discovered rapidly mutating Y-STR markers in UAE population is presented [3]. These markers are DYF387S1, DYF399S1, DYF403S1 and DYF404S1.

2. Materials and methods

2.1. Samples collection and extraction

The UAE population male individuals sample set consisted of 361 buccal swab samples randomly collected from Arabs with informed consent. DNA was extracted using EZ1 extraction robot following manufacturer's protocols and quantified by the Quanti-Fluor™ Human DNA Quantification Kit using a 7500 Real Time System (Applied Biosystems) following the manufacturer's protocol.

2.2. Amplification and detection

Each marker was amplified separately in singleplex reaction for each sample using previously published primers sequences [2] labelled with FAM fluorescent dye. A total reaction volume of 10 µl comprises 5 µl AmpliTaq Gold Master Mix, 0.5 µM of each forward and reverse primers, 0.5ng of DNA template and 3 µl of PCR graded water. Cycles conditions were as the following:

- 95 °C for 5 min.
- 28 cycles of:
 - 94 °C denaturation for 30 s.
 - 60 °C annealing for 30 s.
 - 72 °C extension for 45 s.
- 72 °C final extension for 40 min.

Amplified products were analyzed by capillary electrophoresis by an ABI 3500 Genetic Analyzer for fragment length determination of the products using POP-4™ polymer and LIZ600™ as internal lane DNA standard. Allele assignment was carried out by GeneMapper ID-X software. Samples which showed more than the expected number of alleles at each marker was confirmed to be single source of DNA using Identifiler Plus amplification kit following manufacturer protocol.

2.3. Statistics analysis

All statistics shown in this study were carried out using Microsoft Excel software.

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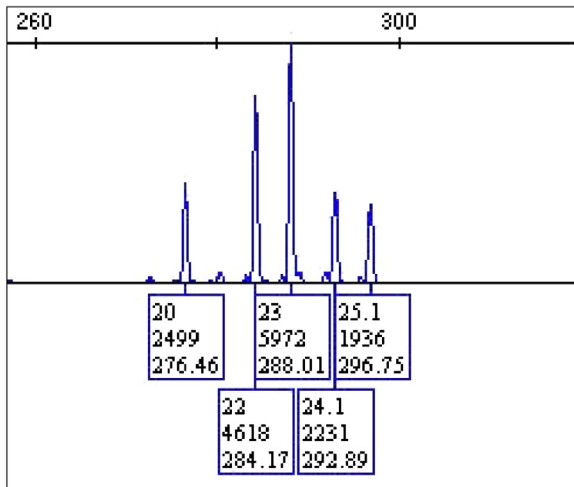


Fig. 1. Alleles called at DYF399S1 marker showing the detection of 8 alleles (E-Type).

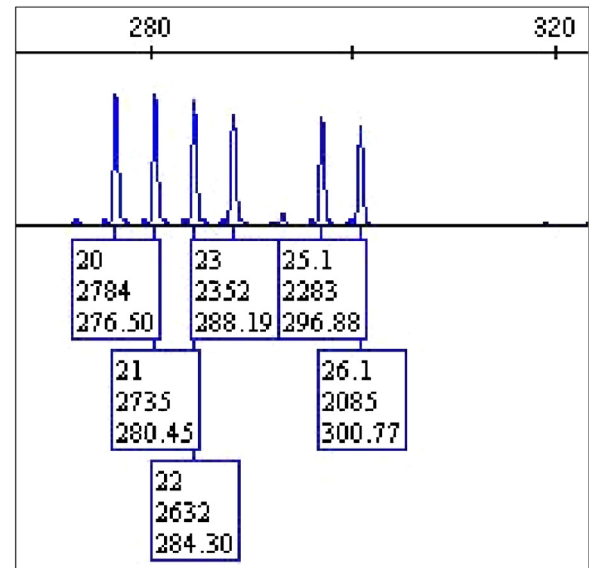


Fig. 2. Alleles called at DYF399S1 marker showing the detection of 6 alleles (E-Type or C-Type).

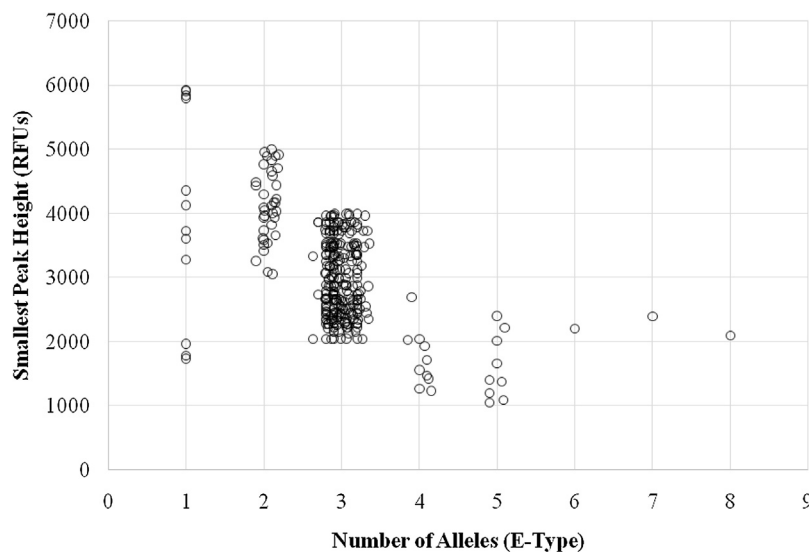


Fig. 3. Consistency in DYF399S1 allele's detection using E-Type genotyping approach for Y-STR analysis.

3. Results and discussion

Out of the four markers, DYF403S1 was excluded from the comparison analysis between the two types of genotyping approaches due to random peak heights observed across the marker and therefore E-Type couldn't be applied for this marker. This result was also confirmed previously [3]. DYF387S1 showed 70 unique genotypes following the C-Type approach and 76 unique genotypes following the E-type approach. DYF399S1 on the other hand, showed 110 unique haplotypes following the C-Type approach and 147 unique haplotypes following the E-Type approach. Interestingly, the highest number of alleles called following the E-type approach was 8 alleles (Fig. 1), whereas the highest number of alleles recorded using the C-type approach was 6 alleles (Fig. 2). A high level of consistency was also detected in

this marker (Fig. 3). Finally, DYF404S1 showed the least haplotype diversity across 361 samples. There were 60 haplotypes detected using the C-Type approach and 62 haplotypes using the E-Type approach.

Conflict of interest

None.

Acknowledgments

We would like to humbly thank Dubai Government for enabling grant support for this project. We also acknowledge Dubai Police General Head Quarters and Dubai Health Authority for helping with collection of population samples.

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