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Evaluation of 13 rapidly mutating Y-STRs on a Dravidian pedigree

S. Iyavoo[a,*], R. Alghafri[b,c], R. Almheiri[b], T. Haizel[a]

[a] Anglia DNA Services, Scottow Enterprise Park, Norwich, UK
[b] General Department of Forensic Sciences and Criminology, Dubai Police General Head Quarters, Dubai, United Arab Emirates
[c] Biology Department, United Arab Emirates University, Al-Ain, United Arab Emirates

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ABSTRACT

The effectiveness of a rapidly mutating Y-STRs multiplex assay (RM Y-STRs) containing 13 RM Y-STR markers was compared with the AmpFℓSTR™ Yfiler™ PCR Amplification Kit (Yfiler) with conventional Y-STR markers for their efficiency in differentiating males within the same paternal lineage. Samples from 4 generations comprising 16 Dravidian males (15 father-son pairs) were analysed with both assays. Mutations were observed in 3 father-son pairs in the RM Y-STRs profiles while only 2 mutations were observed in the Y-filer profiles. Even though not many mutations were observed as anticipated, this study still emphasised the importance of RM Y-STRs when differentiation between males within the same paternal lineage is required and also indicated the need for mutation rates for different populations.

1. Introduction

Y-chromosome short tandem repeats (Y-STRs) profiling is a useful tool for forensic analysis to process challenging cases such as sexual assault when the recovered male DNA is in a small amount compared to the female DNA. However, in cases where the suspects have the same paternal lineage, conventional Y-STR markers would not be very robust to differentiate them. Thus, Y-STR markers with more discriminative power are required to resolve such cases [1].

2. Materials and methods

2.1. Sample collection and DNA extraction

Buccal swabs from 4 generations comprising 16 Dravidian males (15 father-son pairs) within the same paternal lineage were collected with informed consent. DNA from buccal swabs was extracted using the QIAamp® DNA Mini Kit (Qiagen®). DNA quantification was carried out using the Quantifiler™ Human DNA Quantification Kit (Applied Biosystems™) on a 7500 Real-Time System (Applied Biosystems™) following the manufacturer's protocol.

2.2. DNA amplification

Extracted DNA samples were amplified with the AmpFℓSTR™ Yfiler™ PCR Amplification Kit (Applied Biosystems™) and the rapidly mutating Y-STRs multiplex assay. Yfiler amplification was carried out following the manufacturer's recommendation, while a previously published method was used for the RM Y-STRs [2].

2.3. Electrophoresis and typing

Capillary electrophoresis was performed on the ABI Prism® 3500xL Genetic Analyzer (Applied Biosystems™) and alleles were determined using the GeneMapper® ID-X software (Applied Biosystems™).

2.4. Quality controls

Manufacturer provided AmpFℓSTR™ Control DNA 007 and AmpFℓSTR™ Control DNA 9947A were used as the positive and negative controls respectively in Yfiler amplification while previously genotyped controls were used in the RM Y-STRs amplification. The paternity of all father-son pairs were confirmed using the VeriFiler™ Express PCR Amplification Kit (Applied Biosystems™) with a threshold of paternity probability set at 99.99%.

3. Results

In this study, RM Y-STRs amplified 20 alleles compared to 17 in Yfiler. There is no overlapping marker between both assays. The core DNA profiles for this Dravidian pedigree which were developed using both assays are shown in Table 1.

* Corresponding author.
E-mail address: dr_iyavoo@yahoo.com (S. Iyavoo).

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Table 1
Table below shows the core DNA profiles of this Dravidian pedigree for RM Y-STRs and Y-filer loci.

RM Y-STRs locus	Core profile	Y-filer locus	Core profile
DYS526a	13	DYS456	15
DYS612	39	DYS389I	12
DYF399S1	20,21,24.1	DYS390	24
DYS526b	33	DYS389II	30
DYS547	44	DYS458	20
DYF404S1	13,14	DYS19	15
DYS626	29	DYS385	11,14
DYF403S1a	12,13,14	DYS393	14
DYF403S1b	47	DYS391	10
DYS576	19	DYS439	13
DYS518	38	DYS635	22
DYS627	19	DYS392	11
DYS570	19	Y_GATA_H4	12
DYF387S1	38	DYS437	14
DYS449	29	DYS438	11
		DYS448	19

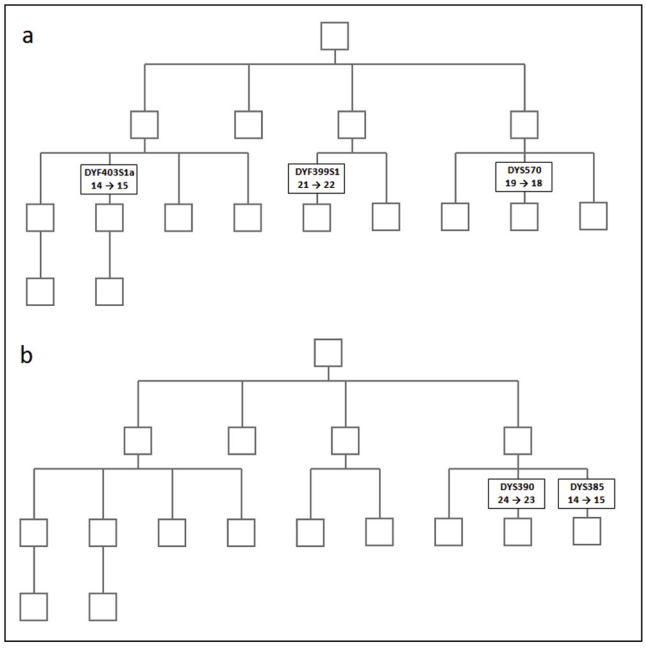

Fig. 1. Pedigrees with observed mutations from RM Y-STRs (a) and Yfiler (b) in 16 Dravidian male samples comprising 15 father-son pairs.

From the total of 15 father-son pairs, mutations were observed in 3 father-son pairs in the RM Y-STRs profiles while only 2 mutations were observed in the Y-filer profiles. All the observed mutations were identified at different loci and were 1-step mutations. Mutations were also observed in the same father-son pair on both the assays and coincidently, both the mutations in this father-son pair were with repeat losses while the remaining 3 mutations were with repeat gains (Fig. 1).

4. Discussion and conclusion

The geographical location of Dravidians is mainly in South India and they belong to the Dravidian linguistic group [3]. The YHRD website (Y-STR Haplotype Reference Database, www.yhrd.org/) comprises 1327 haplotypes (at least minimal) and 13 unique population samples for the Dravidian Metapopulation. The core Yfiler DNA profile from the Dravidian pedigree in this study was searched on this website. However, no match was found even with the minimal haplotype [4] search (accessed on 13th September 2019).

Also based on the YHRD website, the highest combined mutation rate (μ) for the Yfiler loci is at locus DYS458 (μ: 0.00617), followed by locus DYS439 (μ: 0.00546) and locus DYS456 (μ: 0.00441). However, no mutations were identified at these loci. The observed mutations were at locus DYS385 (μ: 0.00251) and locus DYS390 (μ: 0.00208).

According to Ballantyne et al. [1], the highest mutation rate in the RM Y-STRs is at locus DYF399S1 (μ: 0.0773), followed by locus DYF403S1a (μ: 0.031). Mutations at both these loci were also observed in this Dravidian pedigree. Another mutation was observed at locus DYS570 (μ: 0.0144). In a study by Chen et al. [5], the mutation rate at locus DYS570 (μ: 0.0278) was found to be much higher than the mutation rate reported by Ballantyne et al. The samples used in both these studies were from different populations; Chen et al. used samples of Han population from Shandong province, China, while samples used by Ballantyne et al. were obtained from the Berlin, Leipzig, and Cologne areas of Germany and the Warsaw and Wroclaw areas of Poland. Also, only 3 mutations were observed among 15 father-son pairs in this Dravidian pedigree while 17 mutations were identified in 28 father-son pairs in a study of UAE Arabs [2]. These findings indicate that the mutation rates for RM Y-STRs could be different among different populations.

Conclusively, incorporation of the RM Y-STRs based on the mutation rates in different populations could improve the differentiation between males within the same paternal lineage.

Declaration of Competing Interest

None.

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