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Characterization of the STR loci allele's distribution of Y chromosome with high mutation rate in population sample of Rio de Janeiro

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ABSTRACT

Y chromosome genetic markers are used for characterization of male lineages, since they are fully transmitted to next generations unless mutations occur. RM-YSTRs markers display high mutation rates, which is unusually observed in other Y-STRs markers, and seem to be efficient in discriminating paternally related males. The aim of this study was to estimate population and mutational parameters of 13 RM-YSTRs in 258 males born in Rio de Janeiro, Brazil. Population analysis showed high discrimination capacity and no population substructure. Elevated mutation rates were found in this population. For a better characterization of these loci in different Brazilian populations more studies are needed.

1. Introduction

Knowledge about Y chromosome and its genetic markers might be employed at different situations, including evolutional and biogeographic studies, besides forensic and paternal kinship investigations. Genetic Y chromosome markers characterize paternal inheritance, since they are fully transmitted to next generations unless mutations occur [1–3]. Y short tandem repeats markers (Y-STRs) are widely applied in forensic genetics because of their high capacity of discriminate lineages. One of main peculiarities that are sought in markers to be used for this purpose is the high degree of polymorphism, which as a result would offer a more accurate discrimination even between individuals from a unique patrilineal lineage [4,5]. It has been verified elevated mutation rates for several newly described rapidly mutating Y-STR markers (RM-YSTR) and, in consequence, higher polymorphic degree and capacity of discrimination in comparison to other Y-STRs [6,7]. In this study, we aimed to deepen the knowledge about general population data, as well as mutational rates of RM-YSTR markers in Rio de Janeiro, Brazil admixed population.

2. Materials and methods

Blood samples were collected from 258 males born in Rio de Janeiro, Brazil, grouped in 129 fathers/sons pairs, after signing the consent forms by the participants. This study was approved by the Ethics Committee in Research of Pedro Ernesto University Hospital (CAAE: 0067.0.228.000-09). DNA was extracted by the Chelex method. Target sequences were amplified by three polymerase chain reactions (PCR), according to literature protocol [8], and the amplicons were separated through electrophoresis on automated sequencer ABI-3500 (Applied Biosystems). When mutations were detected, they were confirmed by sequencing. The haplotype frequencies, genetic and haplotypic diversities were determined with Arlequin software v3.5 [9]. Mutational rates were determined in Microsoft Excel 2007.

3. Results and discussion

RM-YSTR loci analysis of 129 unrelated individuals showed low haplotype frequencies (0.0077) and haplotype diversity of 1.0 (±0.0010), which is in accordance to the highly polymorphic nature of these markers. Moreover, high values of genetic diversities were obtained for the 13 markers. These results are associated with high mutation rates found (Table 1), with an average rate about 2.11×10^{-2}, which is considered elevated in comparison with the mutation rates of Y-STRs normally used in forensic analyses. In 28 fathers/sons pairs were observed 30 mutations over all studied RM-YSTR loci, excepting DYS518 and DYS626 loci. It was possible to observe that DYF399S1 and DYF403S1a/b multiallelic loci displayed the most

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Table 1
Mutational characteristics of RM-YSTR loci in Rio de Janeiro admixed population.

Locus	Number of mutations	Number of meiosis	Mutation rate
DYF387S1	2	129	1.55×10^{-2}
DYS570	1	129	0.77×10^{-2}
DYS576	2	129	1.55×10^{-2}
DYF399S1	10	129	7.75×10^{-2}
DYS526a/b	2 (0 + 2)	129	1.55×10^{-2}
DYS627	1	129	0.77×10^{-2}
DYS547	1	129	0.77×10^{-2}
DYS449	2	129	1.55×10^{-2}
DYS612	2	129	1.55×10^{-2}
DYF404S1	1	129	0.77×10^{-2}
DYF403S1a/b	6 (5 + 1)	129	4.65×10^{-2}
Mean			2.11×10^{-2}

elevated mutation rates between all of them, which can be attributed to their allelic structure.

All 30 mutations found were single-step, showing 18 repeat gains and 12 repeat losses, which is not generally described in the literature, where it is common to find more repeat losses than gains.

4. Conclusion

Due to their high mutation rates and discrimination capacity, RM-YSTR showed elevated capacity of differentiation in Rio de Janeiro population sample proving to be more discriminative than other Y-STR markers commonly used in population studies and forensic analysis. Therefore, it is possible to conclude that these markers are very promising in discriminate individuals and it will be necessary to analyze other Brazilian populations in order to evaluate and compare populational and forensic parameters.

Conflict of interest

None.

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