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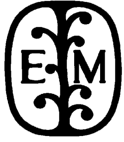

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Y-chromosome STRs in populations of Bantu origin from Mozambique: male contribution to the Africa genetic pool and forensic implications

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Abstract

A set of seven common Y-chromosome specific microsatellites has been used in the present report to type 308 individuals of 16 African population groups from Mozambique. These microsatellites include the DYS19, DYS390, DYS391, DYS392, DYS393, DYS389I and DYS389II systems. The population structure was analysed and therefore genetic distances and several diversity indices were computed and compared with other populations around the world. The results obtained in the present study show that these populations share common characteristics with other available Bantu-speaking African population groups and clear differences with non-African ones. The combination of the analysis of Y-chromosome STRs with other more slowly mutating Y-chromosome polymorphisms (biallelic markers), mtDNA sequences and autosomal data for the same populations, will allow the analysis of human evolution in paternal lineages in different time scales. The forensic implications of these results are discussed.
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Keywords: Y chromosome; STRs; Haplotype diversity; Africa; Mozambique

1. Introduction

Y-chromosome microsatellites (also called short tandem repeats or STRs) provide a potentially valuable system for forensic DNA typing, for evolutionary and genealogical purposes. These markers are highly polymorphic and can be easily studied by using PCR analysis and automated detection. A set of seven common Y-chromosome specific micro-

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satellites has been used in the present study to type 308 individuals of 16 African ethnic groups from Mozambique. These microsatellites include the DYS19, DYS390, DYS391, DYS392, DYS393, DYS389 I and DYS389 II systems. The combination of the analysis of Y-chromosome STRs with other, more slowly mutating, Y-chromosome polymorphisms (biallelic markers), mtDNA sequences and autosomal data for the same populations, will allow the analysis of human evolution in paternal lineages in different time scales. Our aim is to compare the Y-chromosome STR patterns of the south of Africa with those of Caucasoid and Asiatic ones, to know the population behavior of these STRs in populations from different ethnic groups. This population data also have interest for forensic genetics.

2. Materials and methods

2.1. Samples

We analyzed a sample of 308 unrelated healthy men of 16 African population groups from Mozambique: Rongas, Changanes, Tswas, Chopes, Bitongas, Shonas, Ndaus, Nyanjas, Makuas, Lomwe, Chuabos, Yao, Macondes, Nhungwes, Senas and Nguni. DNA was extracted from blood stains as described by Valverde et al. [1]. DNA quantification was performed using fluorescence detection with a DyNAQuant 200 (Amersham Pharmacia Biotech, Uppsala, Sweden). For parts of the data analysis, additional samples from different population groups around the world were included.

2.2. STR polymorphism typing

Six tetranucleotide repeat polymorphism (DYS19, DYS389I, DYS389II, DYS390, DYS391 and DYS393) and one trinucleotide repeat polymorphism (DYS392) were typed in all samples. PCR amplification was carried out in two multiplex reactions: a pentaplex system following the conditions according to Gusmão et al. [2] and a duplex amplification for DYS391 and DYS392, using the primers described in the Genome Database, with the exception of the DYS391, where we used the primers described by Gusmão et al. [3]. Duplex reactions were performed using 25 ng of genomic DNA, 0.12 μM DYS391 primers, 0.2 μM of the DYS392 primers according to González-Neira et al. [4]. The PCR cycling conditions were 30 cycles of 94 °C for 1 min, 58 °C for 1 min and 72 °C for 1 min, with a previous denaturation of 2 min at 95 °C, using a PE thermocycler. For genetic typing, an ABI 377 automated sequencer (Perkin Elmer) along with Genescan 2.1 analysis software was used. Allele designations were based on comparison with the allelic ladders obtained by the mixture of previously sequenced samples for the most common alleles. Allele nomenclature was as proposed by Kayser et al. [5], except for the DYS389 I and II loci, where the nomenclature was according to González-Neira et al. [4].

2.3. Statistical analysis

Allele and haplotype frequencies, gene and haplotype diversity and some Y-chromosome diversity indices were calculated using the ARLEQUIN package version 1.1 [6].

Population differentiation was analyzed with the Markov test also using the ARLEQUIN software. The apportionment of genetic variation within and between populations was estimated by means of AMOVA [7].

3. Results

3.1. Single locus analysis

The gene diversities of each STR for the populations under study were estimated (Table 1). Gene diversities in Y-STRs are lower than those found in autosomal STRs, probably due to the effect of genetic drift, which acts more strongly on the Y chromosome than on autosomes (the effective population size of the Y chromosome is one quarter that of autosomal chromosomes) [8]. DYS391 and DYS392 showed an extremely low gene diversity in all the populations studied. The interpretation of these results should be done in the correct context: the low gene diversity found for these microsatellites in these population groups could be due to an ascertainment bias: a microsatellite selected in a focal population will differ systematically from its orthologues in related populations due to the criteria used to isolate it in the focal species. That is, these Y chromosomes have been originally identified in Caucasoid populations which makes it difficult to compare our results with existing Caucasoid population studies.

3.2. Haplotype analysis

A more consistent population analysis can be performed using haplotypes instead of single locus frequencies. In a total of 308 samples analyzed, 126 different haplotypes were

Table 1
Gene diversity for each locus

	Rongas	Changanes	Tswas	Chopes	Bitongas	Shonas	Ndaus	Nyanjas
DYS19	0.59	0.67	0.46	0.58	0.69	0.61	0.57	0.54
DYS389I	0.60	0.54	0.70	0.62	0.65	0.37	0.61	0.62
DYS389II	0.47	0.72	0.70	0.68	0.68	0.50	0.67	0.57
DYS390	0.34	0.58	0.29	0.62	0.67	0.39	0.65	0.28
DYS391	0.00	0.39	0.46	0.26	0.54	0.39	0.41	0.27
DYS392	0.00	0.00	0.00	0.07	0.19	0.11	0.37	0.10
DYS393	0.68	0.47	0.44	0.40	0.56	0.71	0.61	0.70
	Makuas	Lomwe	Chuabos	Yao	Macondes	Nhungwes	Senas	Nguni
DYS19	0.47	0.72	0.65	0.76	0.60	0.43	0.53	0.41
DYS389I	0.49	0.54	0.49	0.78	0.67	0.49	0.45	0.68
DYS389II	0.66	0.73	0.67	0.60	0.75	0.68	0.71	0.55
DYS390	0.28	0.56	0.28	0.65	0.51	0.28	0.35	0.56
DYS391	0.19	0.36	0.42	0.20	0.41	0.48	0.32	0.00
DYS392	0.10	0.19	0.00	0.20	0.00	0.36	0.00	0.00
DYS393	0.57	0.59	0.70	0.64	0.64	0.60	0.59	0.77

found. The most frequent haplotype for all populations 15-13-18-21-10-11-13 (7%) is present in 22 samples. The average haplotype diversity was 97% (Table 2). The high haplotype diversities in all the populations analyzed reflects the high intrapopulation variability of Y STR haplotypes thus supporting the use of the haplotype system for forensic applications. Moreover, additional data of haplotypes from 66 populations (African and non-African groups) around the world [4,9–12] were used to analyze the shared haplotypes between populations. Comparing the population groups from Mozambique with the other African groups, the highest number of common haplotypes was found to be between the Mozambique ethnic groups and the Ovambos group from Namibia (southeast of Africa). These populations shared a high number of haplotypes with other populations from the south of Africa. However, the lowest number of shared haplotypes was observed between Mozambique population groups and the populations from the north of Africa.

3.3. The proportion of genetic variance through analysis of AMOVA

Three percent of genetic variation was found to be between populations, while the remaining 97.0% was found within populations. The low values of between-population variation found could indicate that these populations are very close to each other, which supports the idea of a common Bantu origin for these population groups. Another AMOVA was performed considering the worldwide sample and comparing African populations with non-African groups. Only 12.7% of the total variation was attributable to differences between population groups, while 15.91% was attributable to differences between populations, within groups. The rest of the variation (71.39%) was attributable to differences within populations.

Table 2
Haplotype diversity by subpopulations

	Rongas	Changanes	Tswas	Chopes	Bitongas	Shonas	Ndaus	Nyanjas
Sample size	21	22	19	27	20	18	19	20
Number of haplotypes	14	15	15	18	16	15	16	16
Polymorphic sites	12	18	15	18	19	19	21	16
Haplotype diversity	0.96±0.03	0.96±0.20	0.96±0.04	0.96±0.02	0.97±0.03	0.97±0.03	0.98±0.02	0.97±0.03
	Makuas	**Lomwe**	**Chuabos**	**Yao**	**Macondes**	**Nhungwes**	**Senas**	**Nguni**
Sample size	20	20	20	10	19	20	21	12
Number of haplotypes	14	16	19	10	16	16	17	12
Polymorphic sites	17	20	19	20	17	18	16	13
Haplotype diversity	0.92±0.05	0.98±0.02	0.99±0.18	1.00±0.05	0.98±0.02	0.96±0.03	0.98±0.02	1.00±0.03

4. Discussion

We have shown in this work preliminary results obtained from the analysis of seven Y-chromosome STRs in 16 population groups from Mozambique, all of them of Bantu origin. We are now studying the matrilineal contribution of the same individuals from these African ethnic groups through the analysis of mitochondrial DNA variation and this information will contribute to knowledge of the genetic landscape of the Bantu-speaking populations. Moreover, the combination of the analysis of Y-chromosome STRs with other, more slowly mutating, Y-chromosome polymorphisms (biallelic markers) will provide a better understanding of human evolution in these paternal lineages at different time scales. Beside this, molecular analysis of Y-chromosome STRs in these ethnic groups has allowed us to know the population behavior of these systems and its potential as tools of forensic usefulness.

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