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Genetic landscape of the mitochondrial DNA control region in South African populations

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ABSTRACT

A high-quality dataset of the mitochondrial DNA control region sequences was generated with Sanger and next-generation sequencing. The study was aimed at characterizing the maternal genetic ancestry and analyzing the haplogroup distribution of 246 individuals residing in South Africa. The study presents mtDNA sequences in South Africa for forensic applications.

1. Introduction

The mitochondrial DNA control region (nucleotide position 16024-576) is valued in forensics when DNA is highly degraded or in cases where maternal lineage information is relevant. The control region is of interest due to its vast majority of rapidly mutating sites [1].

Full mitochondrial DNA control region sequences were obtained from 246 individuals belonging to four population groups in three provinces of South Africa: native Nama (KhoiSan) and Bantu from the Northern Cape province and admixed Coloured and Griqua from the Western Cape (where the two populations originated), Northern Cape and KwaZulu-Natal provinces. Their complex genomic admixture is a result of intermarriages between native KhoiSan inhabitants, individuals from the Bantu expansion and the arrival of the European settlers, SE-Asian and Indian colonies.

The study focused on the contribution of the mtDNA haplotype data, accepting that the enhancement and update of databases is a key objective for the consolidation of the use of mitochondrial DNA for forensic purposes. Therefore, the matrilineal haplotype data will be made available through EMPOP [2] at the end of the study.

2. Methods

Ethical clearance 15-4-97 was granted by the University of the Western Cape Senate Research Committee. Full mitochondrial DNA control region sequences were generated using Sanger method [3] and next-generation sequencing on an Illumina MiSeq platform [4]. The sample-set encompasses 246 individuals belonging to four self-declared ethnic groups in three provinces of South Africa.

Sequences were aligned to the revised Cambridge Reference Sequence [5] to generate haplotype profiles of single nucleotide polymorphisms (SNPs). SNPs were recorded following the nomenclature guidelines set out by the DNA Commission of the International Society for Forensic Genetics [6,7]. The individual profiles were assigned haplogroups using HaploGrep 2 [8]. Haplotype and haplogroup frequency were analyzed with Microsoft Office Excel. Population summary statistics were calculated with DnaSP v5, excluding gaps [9]. Random match probability was calculated as the sum of squares (SS) of the haplotype frequencies.

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Table 1
Forensic and population parameters in four population groups in South Africa.

Statistics	Ethnic group			
	Coloured $n = 138$	Griqua $n = 88$	Bantu $n = 10$	Nama $n = 10$
Haplotypes	63	42	8	8
Unique haplotypes	44	30	6	6
Match probability	0.057	0.039	0.04	0.04
Genetic Diversity	0.943	0.961	0.956	0.956
Nucleotide diversity (π)	0.00542	0.01035	0.01125	0.01436

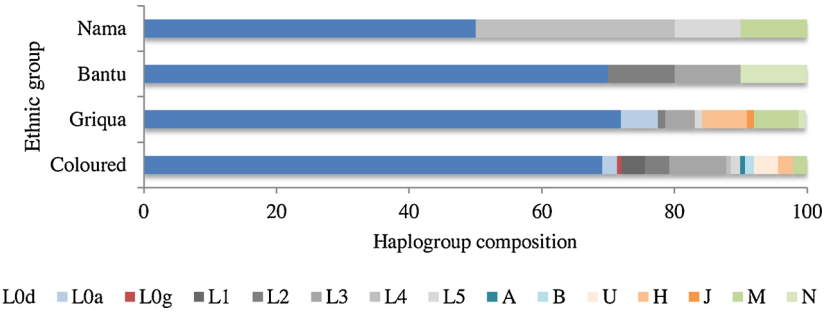

Fig. 1. Mitochondrial haplogroup composition of South African population groups. Haplogroup L0 and L1-5 present KhoiSan and Bantu lineages, respectively. All other haplogroups are of Eurasian ancestry.

3. Results and discussion

A total of 90 mtDNA haplotypes were observed among the 246 individuals residing in South Africa. This includes 61 unique mtDNA haplotypes, while 29 haplotypes were shared between 175 unrelated individuals in the South African sample-set. Table 1 presents a descriptive statistical analysis for each ethnic group in this study.

Table 1 illustrates a high genetic diversity in the Coloured, Griqua, Bantu and Nama ethnic groups in South Africa and Fig. 1 shows the immense L0 maternal haplogroup contribution in all of the population groups.

Fig. 1 illustrates the higher heterogeneity in the admixed Coloured and Griqua population in comparison to the native Bantu and Nama population. Our data revealed higher frequencies for the Native African ancestry and the overall predominance of KhoiSan lineages. All ethnic groups presented evidence of Eurasian ancestry.

4. Conclusion

The high genetic diversity and low random match probability obtained in this investigation demonstrate that the data can be a valuable tool in forensic casework in South Africa. Therefore, the haplotype data will be made available, as it will aid in the assessment of the rarity of mtDNA haplotype frequencies that is essential in forensic and/or missing persons cases.

Declaration of Competing Interest

None.

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