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Research article

mtDNA diversity in Sudan (East Africa)

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Abstract

A major effort must be put in East and sub-Saharan African mtDNA diversity characterisation for the construction of an informative database. We contribute 102 new HVRI + HVRII Sudanese sequences. As expected this sample is highly diverse, mainly constituted of unique haplotypes (2.07% random match probability for HVRI alone), 72.5% of which belong to sub-Saharan haplogroups.

Keywords: mtDNA; Sudan; HVRI; HVRII; Haplotype match

1. Introduction

East African populations present the highest genetic diversity levels, namely for mitochondrial DNA (mtDNA). It is known that the vast majority of mtDNA haplotypes in a population are unique, rendering that mtDNA databases must have a considerable size in order to be informative [1]. The effort to construct African mtDNA databases is still incipient, given the huge sample sizes that will be needed for having forensic discrimination power in such highly diverse populations. Here we report the mtDNA diversity (for hypervariable regions I and II, HVRI and HVRII) in 102 individuals from Sudan, an Eastern African country so far poorly characterised for mtDNA diversity.

2. Material and methods

A total of 102 unrelated Sudanese were sampled. MtDNA was amplified using the primers L15997 (5′-CAC CAT TAG CAC CCA AAG CT-3′) and H16401 (5′-TGA TTT CAC GGA GGA TGG TG-3′) for HVRI and L48 (5′-CTC ACG GGA GCT CTC CAT GC-3′) and H408 (5′-CTG TTA AAA GTG CAT ACC GCC A-3′) for HVRII. The temperature profile was 95 °C for 10 s, 60 °C for 30 s and 72 °C for 30 s, for 35 cycles of amplification. The amplified samples were purified with Microspin™ S-300 HR columns (Amersham Biosciences), according to the manufacturer's specifications. The sequence reactions were carried out using the kit Big-Dye™ Terminator Cycle Sequencing Ready Reaction (AB Applied Biosystems), with one of the above primers, in both forward and reverse directions. Sephadex 50 was used for post-sequence reaction purification of samples, which were then run in an automatic sequencer ABI 3100 (AB Applied Biosystems).

The nucleotide positions considered for the analysis of the sample were 16024–16383 for HVRI and 73–340 for HVRII. Length variation (often scored as transversions in HVRI) was not considered [2]. Sequence classification into haplogroup was done according to Ref. [3]. Molecular diversity indexes and AMOVA were calculated in Arlequin 2.0 [4]. The random match probability was calculated as the sum of the square of haplotype frequencies. The following samples were used for comparison: Egypt ($n = 68$) [5]; Nubia ($n = 80$) [5]; Ethiopia ($n = 270$) [6]. As these populations were not surveyed for HVRII diversity, calculations were applied to HVRI alone, but HVRII information was taken into account for haplogroup affiliation.

3. Results

As expected, levels of diversity were high, attaining a value of 0.989 ± 0.004 for haplotype diversity in HVRI, comparable

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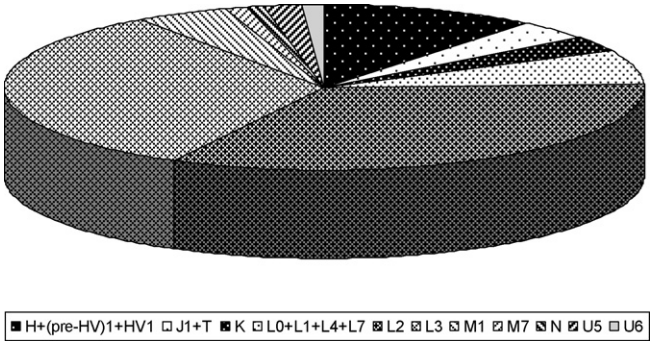

Fig. 1. Haplogroup distribution (%) in Sudan.

with values in neighbouring populations: 0.993 ± 0.005 in Egypt; 0.977 ± 0.008 in Nubia; 0.993 ± 0.001 in Ethiopia. The random haplotype match probability in HVRI was of 2.07% in Sudan, 2.16% in Egypt, 3.54% in Nubia and 1.06% in Ethiopia.

The AMOVA results for these Eastern African samples, considered as a group, indicated that most of the diversity is present inside populations (98.89% of variation).

The haplogroup distribution in Sudan (Fig. 1) was: 22.5% of Eurasian ancestry; 4.9% of the East African M1 lineage; 72.5% of sub-Saharan affiliation. In the sub-Saharan pool, a proportion of 44.6% is represented by the haplogroup L3, the ancestor of the worldwide mtDNA diversity outside Africa.

4. Conclusions

The African mtDNA diversity characterisation needs a clear wider survey. As already known, and showed here again, populations are largely constituted by unique haplotypes, rendering that almost all diversity is present inside populations.

The 102 new HVRI + HVRII Sudanese sequences presented here belong mainly to sub-Saharan haplogroups (72.5%), with lesser proportions of Eurasian (22.5%) and typical East African M1 haplogroup (4.9%) backgrounds.

Given the recent interest in the alternative routes for out-of-Africa migration(s) (Levant versus Southern), the L3 haplotypes, in the root of all non-African diversity, will contribute information to shed light on this issue.

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Conflict of interest

None.

References

[1] L. Pereira, C. Cunha, A. Amorim, Predicting sampling saturation of mtDNA haplotypes: an application to an enlarged Portuguese database, Int. J. Legal Med. 118 (2004) 132–136.
[2] K.E. Bendall, B.C. Sykes, Length heteroplasmy in the first hypervariable segment of the human mtDNA control region, Am. J. Hum. Genet. 57 (1995) 248–256.
[3] A. Salas, M. Richards, T. De la Fe, et al., The making of the African mtDNA landscape, Am. J. Hum. Genet. 71 (2002) 1082–1111.
[4] S.D. Schneider, D. Roessli, L. Excoffier, A Software for Population Genetics Data Analysis, University of Geneva, 2000 (Arlequin Version 2.000).
[5] M. Krings, A.E. Salem, K. Bauer, H. Geisert, A.K. Malek, L. Chaix, C. Simon, D. Welsby, A. Di Rienzo, G. Utermann, A. Sajantila, S. Paabo, M. Stoneking, mtDNA analysis of Nile River Valley populations: a genetic corridor or a barrier to migration? Am. J. Hum. Genet. 64 (1999) 1166–1176.
[6] T. Kivisild, M. Reidla, E. Metspalu, A. Rosa, A. Brehm, E. Pennarun, J. Parik, T. Geberhiwot, E. Usanga, R. Villems, Ethiopian mitochondrial DNA heritage: tracking gene flow across and around the gate of tears, Am. J. Hum. Genet. 75 (2004) 752–770.