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Mitochondrial analysis of a British Afro-Caribbean population

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Abstract. Mitochondrial DNA (mtDNA) analysis has been used both to identify individuals and to infer patterns of human evolution. We have analysed a representative population of Afro-Caribbean individuals who are residents within the UK. Sequencing has been carried out on both the hypervariable regions I (HVI) and II (HVII). In addition, a set of informative mitochondrial single nucleotide polymorphisms (SNPs) have been investigated. Comparison of these results with published data for different African populations has enabled the samples to be classified into haplogroups. Analysis of the haplogroup distribution has suggested an ancestral geographical origin for these British Afro-Caribbean samples mainly from Western Africa with some influence from Southeast Africa.

Keywords: Mitochondrial DNA; British Afro-Caribbean population

1. Introduction

Mitochondrial DNA (mtDNA) analysis is becoming more widely used in forensic science, especially in cases of sample degredation and complex relationship issues. It is also proving to be a useful tool in the study of recent human evolution.

There are two areas of the mtDNA genome that are normally sequenced in forensic analysis, hypervariable regions I (HVI) and II (HVII). To interpret the results, the haplotypes obtained need to be compared against a representative database.

Migration into Britain has led to a multicultural society, with 2.2% of the population classing themselves as Black by the 2001 census [1]. A database of British Afro-Caribbeans was therefore established to allow assessment of haplotype frequency in cases involving families or individual from this racial group. By including some informative mitochondrial single nucleotide polymorphisms (SNPs), it is possible to further define these samples into

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mitochondrial haplogroups. This enables a general picture to be built up regarding the composition of a British Afro-Caribbean population in relation to their African origins.

2. Method

DNA samples were obtained from 128 unrelated individuals resident in the United Kingdom who describe their ethnic origin as Black. Samples had previously been used for paternity testing and were randomly selected. Consent was obtained in all cases.

Following Chelex extraction, HVI and HVII were each amplified directly via PCR. The PCR products were incubated with ExoSAP-IT (Amersham Biosciences) to remove unincorporated primers and dNTPs and sequenced using the DYEnamic ET Terminator Cycle Sequencing kit (Amersham Biosciences). The same primers were used as in the initial PCR. In cases where length heteroplasmy was observed in the cytosine stretch from 16184 to 16193, alternative primers that bind to the region just downstream were used to provide additional sequencing data.

The sequence reaction products were precipitated with ethanol and sodium acetate/EDTA prior to separation via capillary electrophoresis on an ABI PRISM 3100 Genetic Analyser (Applied Biosystems). Sequence Analysis and Sequence Navigator software (Applied Biosystems) were used to compare the results obtained with the Anderson reference sequence [2], and any deviations from this were noted. The haplotypes produced could then be recorded to provide haplotype frequency information.

A mtDNA skeleton of African haplogroups derived by phylogenetic analysis (detailed by Salas et al. [3]) was used as a guide to classify samples based on the sequence information of HVI and HVII. In cases where the sequence information alone was insufficient for haplogroup classification, mtSNPs in the coding region were additionally typed.

Seven SNPs were selected for the ability to differentiate badly characterized African haplotypes. These were (with reference to the Anderson sequence):

3693—To distinguish haplogroup L2d
8701—To distinguish haplogroup L*
8618—To distinguish haplogroup L3d
10086—To distinguish haplogroup L3b
10400—To distinguish haplogroup M*
10819—To distinguish haplogroup L3e
12705—To distinguish haplogroup N*

Primers were designed and five products were amplified from two PCR reactions. The seven SNPs were typed using SNaPshot (Applied Biosystems) in one multiplex reaction. The SNaPshot products were separated by size on an ABI PRISM™ 3100 Genetic Analyser and GeneScan 3.1 software (Applied Biosystems) was used for assessing allelic designation.

3. Results

Of the 128 samples analysed, 93 different HVI haplotypes and 80 different HVII haplotypes were observed. When HVI and HVII were analysed together, this figure increased to 120 (a haplotype diversity value of 0.9990).

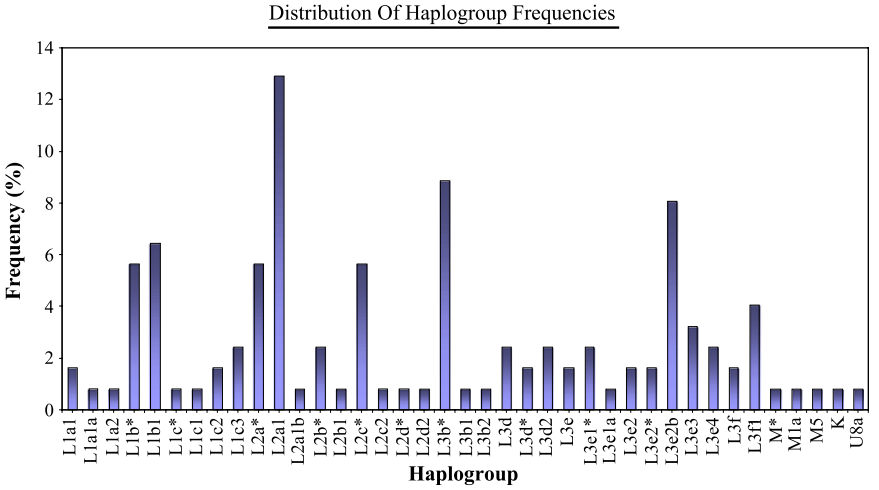

Fig. 1. Haplogroup frequency for British Afro-Caribbean samples.

The haplogroup classifications are represented graphically in Fig. 1. The most common haplogroups were L1b* (5.5%), L1b1 (6.5%), L2a* (5.5%), L2a1 (13%), L2c* (5.5%), L3b* (9%) and L3e2b (8%).

4. Discussion

As expected, the database of British Afro-Caribbeans shows a relatively high diversity. In most cases, the classification of the samples seem to generally point to an origin from West Africa (with some influence from Southeast Africa), whether through direct migration to Britain or via the Caribbean. While there are still large areas of Africa uncharacterized, this proposed distribution occurs despite the cultural and commercial links that Britain has with the Southern and Eastern African countries in the British Commonwealth and so may partly reflect the influence of the Atlantic slave trade.

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