Y Chromosomal SNP Analysis Using the Minisequencing Strategy in a Moroccan Population Samples

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Abstract

The Y chromosome contains the largest non-recombining portion in the human genome. Y-Binary polymorphisms, also known as a single nucleotide polymorphisms (SNPs), are a series of biallelic polymorphisms occurring on the non-recombining region of the Y chromosome (NRY), which represent a precious tool for human evolutionary studies and, potentially, for forensic applications. Low mutation rate, paternal inheritance, and absence of recombination make Y-SNPs particularly suitable for the identification of stable paternal lineages and the reconstruction of an ancestral state from which to explore the evolution of humans. Also, these markers would allow inference of the paternal ancestry of unknown samples which could be useful in forensic applications. In the present study we analyzed 22 biallelic Y-SNPs in 159 males belonging to three ethnic groups (Arab n = 42, Berber n = 67 and Sahrawi n = 50) from Morocco. A total of 10 different haplogroups were identified in this sample representative of Moroccan population. The most common Y chromosome haplogroups is E1b1b1, E1b1b1b and J1 with frequencies of 56%, 49% and 10% respectively.

Keywords: Y-chromosomal SNP; Mini-sequencing; SNaPshot; Morocco

Introduction

Biallelic markers, such as single nucleotide polymorphisms (SNPs) and insertion/deletions (indels), represent an important class of markers on the Y-chromosome [1]. Y-chromosome SNPs (Y-SNPs) are mostly used in molecular anthropology for evolutionary research. Moreover, typing a sample’s Y chromosome haplogroup allows paternal ancestry inference. This may be useful, in forensic applications, when a conventional Short Tandem Repeat (STR) profile, generated from DNA collected at a crime scene, does not match any of the identified suspects and doesn’t “hit” any profile on the available databases.

The kingdom of Morocco is a country located in the northwestern corner of the African continent with coasts on the Atlantic Ocean and the Mediterranean Sea; it is bordered by Algeria to the east and Mauritania to the south. Modern-day Morocco is inhabited by three major ethnic groups (Arab, Berber and Sahraoui). Several dialects such as Arabic (Moroccan dialect or Darija), Berber (Tarifit, Tachelhit and Tamazight) and Sahraoui (El Hassania) are spoken in the country. The aim of the present study was to develop an assay to genotype a selected panel of Y-SNPs using single base extension assay (SBE), or minisequencing [2], in order to determine the most frequent Y-chromosomal haplogroup in Moroccan population. The analysis of single nucleotide polymorphisms (SNPs) is a promising application in forensic casework; since the forensic scientists is often faced with degraded and/or very low amounts of DNA.

Materials and Methods

Population

Buccal swabs were collected from 159 unrelated healthy adult men belonging to the three ethnic groups (Arab n = 42, Berber n = 67 and Sahraoui n = 50). Informed consent was obtained from all participants in this study, and information about the geographical origin of their grand-parents and about their first language was recorded.

DNA Isolation

Genomic DNA was extracted from buccal swab punches, using DNA IQ™ System (Promega; Madison, Wisconsin) on the Biomek® 2000 (Beckman Coulter, Brea, CA) robotic platform according to manufacturer’s instructions.

Y-SNP selection and multiplex design

Assay design and development together with sample testing were conducted in the Forensic Molecular Biology Laboratory of the Forensic Sciences Department of The George Washington University. A total of 22 Y chromosome biallelic markers were selected for this study following a hierarchical strategy based on the phylogenetic tree of Y chromosome recognized by the Y Chromosome Consortium (YCC) [3]. Loci Nomenclature is based on Karafet et al. [4]. The reference sequence for each Y-SNP was taken from the International Society of Genetic Genealogy, Y-DNA Haplogroup Tree 2010 ([http://www.isog.org/tree]). The 22 markers were genotyped in two heptaplex, one hexaplex, and one duplex PCR reactions. The Multiplex MY01 allows the detection of major clades (A-R), A-M91 (A), B-M60 (B), C-RPS4Y (C), D-M174 (D), J-M267 (J1), J-M170 (J) and R-M207 (R). The Multiplexes MY02 E-P147 (E1), E-M132 (E1a), E-P189 (E1b1a), E-M215 (E1b1b), E-M81 (E1b1b1b), E-M54 (E2b), J-M172 (J2), MY03 J-L24 (J2a4h), J-M221 (J2b), R-L63 (R1a), R-M343 (R1b), R-M269 (R1b1b2),

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Received December 16, 2010; Accepted December 29, 2010; Published December 30, 2010


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Nevertheless, this haplogroup (E1b1b1b) is not found in sub-Saharan Africa and its frequency sharply declines through the continent towards the east.

The J1 haplogroup, defined by the single nucleotide polymorphism (SNP) M267, is most frequent in the Arabian Peninsula especially in Yemen (76%) [11]. This could be attributed to the early medieval period during which the Semitic expansion spread J1 out of Arabia into North Africa [12].

The multiplex assays described herein were designed to explore the shallowest branches of Y chromosome haplogroups in Moroccan population. They could also be applied to human evolution and human genetists studies as well as to forensic casework for ancestry inferences.

**Acknowledgement**

We would like to express our sincere gratitude to the Moroccan-American Commission for Educational and Cultural Exchange (MACCEC) and the Fulbright Program for the Fulbright scholarship, for supporting a part of our research in the United States. We would like, also, to thank the George Washington University and the Armed Forces DNA Identification Laboratory (AFDIL). We are greatly indebted to Kimberly Andreaggi Sturk, Jodi A. Irwin, Joni Johnson and Katherine Butler for discussion and technical assistance. Finally, an honorable mention goes to the support offered by GENOME Biotechnologies.

**Reference**