

Announcement of Population Data

Haplotype diversity in human mitochondrial DNA hypervariable regions I–III in the city of Caracas (Venezuela)

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Abstract

In order to expand the database of variable DNA for forensic identification purposes in Venezuela, we analyzed the sequence polymorphisms of mitochondrial DNA (mtDNA) hypervariable regions (HVR) I–III from 100 unrelated individuals from the city of Caracas, using PCR amplification and fluorescent-based capillary electrophoresis sequencing method. Dominant haplogroups corresponded to Native Americans followed by African ones. The inclusion of HVR III although useful for sub-haplogroup assignation, added little to the discrimination capacity of our database. © 2008 Elsevier Ireland Ltd. All rights reserved.

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Population: One hundred healthy unrelated individuals living in the city of Caracas in the north-central region of Venezuela were recruited from undergraduate students and professionals from the Central University of Venezuela and our Institute IDEA. Informed consent was required, and all subject-based procedures were approved by the Venezuelan Bioethics committee from the *Fondo Nacional de Investigaciones Científicas y Tecnológicas* (FONACIT).

DNA extraction: Subjects' fingers were pierced and single blood drops were collected on FTA[®] cards (Gibco-BRL) and processed with the FTA[™] Purification Reagent from Gibco-BRL following the manufacturer's recommendations.

Amplification and sequencing of mtDNA: The following sets of PCR primers were used to amplify three DNA fragments corresponding to HVR I (476 bp), HVR II (361 bp) and HVR III (436 bp): **L15926** 5'-TCA AAG CTT ACA CCA GTC TTG TAA ACC-3', **H16401** 5'-TGATTT CAC GGA GGA TGG TG-3', **L048** 5'-CTC ACG GGA GCT CTC CAT GC-3', **H408** 5'-CTG TTA AAA GTC CAT ACC GCC A-3', **L184** 5'-CGC ACC TAC GTT CAA TAT TAC AG-3', and **H619** 5'-GGT GAT GTG AGC CCG TCT AA-3'. 'L' and 'H' designate the light and the

heavy strands of the mtDNA molecule, respectively. L184 and H619 are primers modified from Lutz et al. [1]. Each PCR was performed using Taq Platinum (Invitrogen[™]) and 30 amplification cycles in a PTC-200 thermocycler device (MJ Research[®]). PCR products were purified using Concert[®] kit (Gibco-BRL) and both strands sequenced using the same primers as for the amplification, and BigDye[™] Terminator v.3.1 Kit (Applied Biosystems) on an ABI Prism[™] 310 Genetic Analyzer. Sequence edition was performed using DNA for Windows software and polymorphisms were analyzed with DNAMAN v.5.2.2 Software, (Lynnon BioSoft). The treatment of length variants was done following recommendations of Bandelt and Parson [2]. The haplogroup assignments were confirmed using the program mtDNAMAN (http://mtmanager.yonsei.ac.kr).

Quality control: Proficiency testing of the GEP-ISFG WG (http://www.gep-isfg.org).

Analysis of data: The probability of randomly selecting two samples with the same mitochondrial profile (random match probability) was calculated using the equation $p = \sum x^2$, where x is the frequency of each mtDNA haplotype [3]. Genetic diversity was estimated using the algorithm $h = n(1 - \sum x^2)/(n - 1)$, where n is the sample size and x is the frequency for each mtDNA haplotype [4]. The rest of statistical parameters were calculated using Arlequin Version 2.000 Software [5].

Results: The mitochondrial haplotypes found are shown in Table 1, whereas in Table 2 the statistical parameters of the database are presented.

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Table 1
mtDNA haplotypes found in Caracas population

HT	Hg	HVI (16024–16365)	HVII (073–340)	HVIII (438–574)	N
V001	A*	16092C 16155G 16223T 16290T 16319A	73G 146C 152C 153G 182T 235G 263G 309.1C 315.1C 316A	523d 524d	1
V002	A2	16111T 16223T 16290T 16319A 16362C	73G 146C 153G 235G 263G 309.1C 315.1C	523d 524d	10
V003	A2	16111T 16223T 16290T 16319A 16362C	73G 146C 153G 235G 263G 309.1C 309.2C 315.1C	523d 524d	3
V004	A2	16111T 16129A 16223T 16290T 16319A 16362C	73G 146C 153G 235G 263G 309.1C 309.2C 315.1C	523d 524d	3
V005	A2	16111T 16223T 16290T 16311C 16319A 16362C	73G 103A 146C 153G 235G 263G 309.1C 309.2C 315.1C	523d 524d	2
V006	A2	16111T 16223T 16290T 16319A 16362C	73G 146C 153G 235G 263G 309.1C 315.1C	456T 523d 524d	1
V007	A2	16111T 16223T 16290T 16319A 16362C	73G 146C 153G 235G 263G 315.1C	485C 523d 524d	1
V008	A2	16111T 16223T 16239T 16284G 16290T 16319A 16362C	73G 146C 153G 235G 263G 309.1C 315.1C	523d 524d	1
V009	A2	16111T 16129A 16223T 16290T 16319A 16362C	73G 146C 153G 235G 263G 315.1C	471C 523d 524d	1
V010	A2	16111T 16223T 16290T 16296T 16319A 16362C	73G 146C 153G 167T 235G 263G 315.1C	523d 524d	1
V011	A2	16111T 16223T 16290T 16319A 16362C	73G 146C 204C 235G 263G 266C 309.1C 315.1C	523d 524d	1
V012	A2	16111T 16129A 16223T 16290T 16293C 16319A 16362C	73G 146C 153G 235G 263G 309.1C 309.2C 315.1C	523d 524d	1
V013	A2	16111T 16223T 16258C 16290T 16319A 16362C	73G 146C 153G 215G 235G 263G 309.1C 309.2C 315.1C	485C 523d 524d	1
V014	A2	16111T 16129A 16223T 16290T 16319A 16362C	73G 146C 153G 235G 263G 309.1C 315.1C	523d 524d	1
V015	A2	16111T 16223T 16290T 16319A 16362C	73G 146C 153G 195C 235G 263G 309.1C 315.1C		1
V016	A2	16111T 16213A 16223T 16290T 16319A 16356C 16362C	73G 146C 153G 235G 263G 315.1C	523d 524d	1
V017	A2	16094C 16111T 16129A 16223T 16290T 16319A 16362C	73G 146C 235G 263G 309.1C 309.2C 315.1C	523d 524d	1
V018	A2	16086C 16111T 16223T 16242T 16290T 16319A 16362C	73G 146C 153G 235G 263G 309.1C 315.1C	523d 524d	1
V019	A2	16111T 16223T 16290T 16319A 16359C 16362C	73G 146C 153G 235G 263G 315.1C	523d 524d	1
V020	A2	16111T 16126C 16223T 16256T 16290T 16319A 16362C	73G 146C 153G 235G 263G 315.1C	523d 524d	1
V021	B*	16166G 16183C 16189C 16193.1C	73G 131C 183G 263G 309.1C 315.1C	499A	1
V022	B4b	16183C 16189C 16193.1C 16217C	73G 158A 263G 315.1C	499A 524.1A 524.2C	1
V023	B4b	16183C 16189C 16193.1C 16217C 16221T	73G 263G 309.1C 309.2C 315.1C	499A 524.1A 524.2C 573.1C	1
V024	B4b	16166G 16167T 16183C 16189C 16193.1C 16217C 16361A	73G 131C 183G 263G 309.1C 309.2C 315.1C	499A	1
V025	B4b	16183C 16189C 16193.1C 16217C 16234T	73G 146C 263G 315.1C	498d 499A	1
V026	C*	16223T 16298C 16327T	73G 146C 152C 249d 263G 309.1C 315.1C	489C 523d 524d	1
V027	C1	16223T 16292T 16298C 16325C 16327T 16362C	73G 249d 263G 290d 291d 309.1C 315.1C	489C 493G 523d 524d	4
V028	C1	16051G 16129A 16223T 16325C 16327T	73G 151T 194T 249d 263G 290d 291d 309.1C 315.1C	489C 523d 524d	2
V029	C1	16223T 16298C 16325C 16327T	73G 115d 249d 263G 290d 291d 309.1C 315.1C	489C	2
V030	C1	16075C 16223T 16298C 16325C 16327T	73G 115d 119C 249d 263G 290d 291d 309.1C 315.1C	489C	1
V031	C1	16051G 16189C 16223T 16298C 16325C 16327T	73G 194T 249d 263G 290d 291d 315.1C	489C 523d 524d	1
V032	C1	16051G 16129A 16223T 16325C 16327T	73G 151T 194T 249d 263G 290d 291d 315.1C	489C 523d 524d	1
V033	C1	16223T 16298C 16325C 16327T	73G 185A 249d 263G 290d 291d 293C 294C 315.1C	489C	1
V034	C1	16223T 16292T 16298C 16311C 16325C 16327T 16362C	73G 152C 249d 263G 290d 291d 309.1C 309.2C 315.1C	489C 493G 523d 524d	1
V035	C1	16223T 16298C 16299G 16325C 16327T	73G 249d 263G 290d 291d 309.1C 315.1C	489C 493G 523d 524d	1
V036	C1	16223T 16271C 16292T 16298C 16325C 16327T 16362C	73G 249d 263G 290d 291d 309.1C 309.2C 315.1C	489C 493G 523d 524d	1
V037	C1	16223T 16292T 16298C 16325C 16327T 16362C	73G 249d 263G 290d 291d 309.1C 309.2C 315.1C	489C 493G 523d 524d	1

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