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Original Communication

Mitochondrial DNA sequence analysis of a native Bolivian population

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ABSTRACT

Mitochondrial DNA analysis is very useful for the interpretation of the history of human migration and to estimate the frequency of a haplotype in the forensic context. From a human settlement perspective, La Paz area is greatly interesting since the first planned city of the region is located there. Samples from 110 individuals from La Paz were studied analysing the polymorphisms in the D-loop, hypervariable region I (HVI) and hypervariable region II (HVII) in order to verify the genetic diversity. The aim of this study was to start the creation of a population database in order to obtain the genetic interpopulation variability and classify haplotypes into characteristic haplogroups of South America. A total of 97 different haplotypes were identified, 90 being unique, expressed by 122 polymorphic nucleotide positions. Nucleotide and sequence diversity were estimated to be 0.015 ± 0.0075 and 0.996, respectively. Haplogroup distribution in the samples was 57.27% B4, 19.09% C1, 10.00% A2, 3.64% D1, 2.73% D4h3, 1.82% H, and 0.91% for each of the haplogroups A4, B4c1a, CZ, D4J, M7a and M8/N9b. The rate of length heteroplasmy was 36.36% in HVI and 52.73% in HVII. Phylogenetic analysis reveals proximity to the Korean, Chilean aboriginal, Japanese and Australian populations. The estimated genetic variability of the studied population was high, suggesting an early settlement.

1. Introduction

Bolivia is a South American country in the heart of the Andes. The history of the territory follows the cultures of the first empire within the domains of the first planned city of the region, Tiwanaku, sited in the South coast of the Lake Titicaca. The last continent to be populated by humans was the Americas and several questions remain open regarding this event. Due to its antiquity, the Andean region can reveal some of the human migration and colonization history of the New World since most of the Bolivian population descends from Aymaras and Quechuas, two cultural groups from the Tiwanaku and Inka Empire, which persisted labouring the fields of the Andean highlands around Lake Titicaca until the present days.[1]

Mitochondrial DNA (mtDNA) is an excellent tool for population studies and forensic genetics due to its high copy number per cell, maternal inheritance,[2] high mutation rate of evolution that provide highly polymorphism to the sequences and absence of recombination.[3] The discrimination power of mtDNA is due to the polymorphic nature of the hypervariable regions (HVI, HVII, and HVIII) which are located in the control region. The mitochondrial genome has become the most widely used genetic marker in human evolution studies. Sequences with shared mutations that tend to reveal a regional specificity define haplogroups. These haplogroups are important to clarify the history and demographic past of a population since they can reflect a phylogenetic relationship between populations.[4] The first mtDNA haplogroups discovered in Native Americans were determined by Torroni et al., 1993.[5] Posterior studies of mtDNA variation concluded that Native American populations have five distinct major haplogroups, all of Asian origin (A, B, C, D and X).[5,6] For forensic purposes, the discrimination power between individuals based on mtDNA is lower than traditional nuclear DNA markers because of its maternal inheritance; however, mtDNA analysis is particularly useful when there are degraded or low level DNA samples. To solve cases of missing persons and to

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Table 1
HVI and HVII mtDNA sequences of 110 individuals from La Paz. N – number of occurrence of the haplotype; Est. Hg – "estimated haplogroup".

	HVI	HVII	N	Est. Hg
LPAZ001	16183C; 16189C; 16217C[a]	73G; 207A; 263G; 315.1C	1	B4
LPAZ002	16223T; 16292T; 16325C; 16362C	73G; 263G; 315.1C	1	D1
LPAZ003	16183C; 16189C; 16217C[a]	73G; 103A; 263G; 315.1C	1	B4
LPAZ004	16093C; 16183C; 16189C; 16217C[a]	73G; 263G; 309.1C; 315.1C	1	B4
LPAZ005	16183C; 16188T; 16189C; 16217C	73G; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ006	16183C; 16188T; 16189C; 16217C	73G; 185A; 263G; 309.1C; 315.1C	1	B4
LPAZ007	16183C; 16188T; 16189C; 16217C	73G; 241G; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ008	16183C; 16189C; 16217C[a]	73G; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ009	16223T; 16325C; 16362C	73G; 143A; 263G; 309.1C; 315.1C	1	D1
LPAZ010	16183C; 16189C; 16217C[a]	73G; 146C; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ011	16183C; 16189C; 16217C; 16270T; 16278T[a]	73G; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ012	16183C; 16188T; 16189C; 16217C; 16362C	73G; 152C; 186T; 263G; 315.1C	1	B4
LPAZ013	16182C; 16183C; 16189C; 16217C[a]	73G; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ014	16183C; 16189C; 16217C[a]	73G; 143A; 146C; 215G; 263G; 315.1C	1	B4
LPAZ015	16168T; 16183C; 16189C; 16217C; 16270T; 16278T[a]	73G; 263G; 315.1C	1	B4
LPAZ016	16223T; 16241G; 16304C; 16325C; 16362C	73G; 263G; 309.1C; 315.1C	1	D1
LPAZ017	16168T; 16182C; 16183C; 16189C; 16217C; 16218T[a]	73G; 263G; 315.1C	1	B4
LPAZ018	16183C; 16188T; 16189C; 16217C	73G; 151T; 186T; 204C; 263G; 315.1C	1	B4
LPAZ019	16182C; 16183C; 16189C; 16217C; 16362C[a]	73G; 263G; 309d; 315.1C;	1	B4
LPAZ020	16183C; 16189C; 16217C; 16293C[a]	73G; 204C; 207A; 263G; 315.1C[a]	1	B4
LPAZ021	16183C; 16189C; 16217C[a]	73G; 151T; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ022	16183C; 16188T; 16189C; 16217C; 16354T	73G; 186T; 263G; 309.1C; 315.1C	1	B4
LPAZ023	16183C; 16189C; 16217C[a]	73G; 143A; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ024	16183C; 16188T; 16189C; 16217C; 16266T	73G; 186T; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ025	16183C; 16188T; 16189C; 16217C; 16354T; 16362C	73G; 186T; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ026	16183C; 16189C; 16217C[a]	73G; 103A; 143A; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ027	16183C; 16189C; 16217C; 16261T; 16319A[a]	73G; 75A; 263G; 315.1C; 340T[a]	1	B4
LPAZ028	16129A; 16183C; 16189C; 16217C[a]	73G; 146C; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ029	16183C; 16189C; 16217C; 16289G[a]	73G; 143A; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ030	16183C; 16189C; 16217C; 16356C; 16362C[a]	73G; 204C; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ031	16183C; 16189C; 16217C[a]	73G; 207A; 257G; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ032	16183C; 16189C; 16217C; 16359C[a]	73G; 143A; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ033	16183C; 16188T; 16189C; 16217C; 16354T	73G; 146C; 186T; 263G; 309.1C; 315.1C	1	B4
LPAZ034	16223T; 16255A; 16301T; 16342C; 16362C	73G; 152C; 263G; 309.1C; 315.1C[a]	1	D4h3
LPAZ035	16183C; 16189C; 16217C; 16360T[a]	73G; 143A; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ036	16183C; 16189C; 16217C; 16274A; 16319A; 16362C[a]	73G; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ037	16223T; 16266T; 16298C; 16325C; 16327T	73G; 249d; 263G; 290d; 291d; 315.1C	1	C1
LPAZ038	16105K; 16168T; 16183C; 16189C; 16217C; 16301A[a]	73G; 204C; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ039	16183C; 16189C; 16217C[a]	73G; 143A; 146C; 215G; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ040	16183C; 16188T; 16189C; 16217C; 16304C; 16362C	73G; 152C; 186T; 263G; 309.1C; 315.1C	1	B4
LPAZ041	16066G; 16183d; 16186T; 16189C; 16217C	73G; 143A; 210G; 263G; 309.1C; 315.1C	1	H
LPAZ042	16114G; 16129A; 16222T; 16223T; 16325C; 16362C	73G; 143A; 263G; 309.1C; 315.1C	1	D1
LPAZ043	16183C; 16189C; 16217C; 16289G[a]	73G; 143A; 146C; 215G; 263G; 309d; 315.1C	1	B4
LPAZ044	16051G; 16182C; 16183C; 16189C; 16217C[a]	73G; 152C; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ045	16183C; 16189C; 16217C; 16301A[a]	73G; 146C; 197G; 215G; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ046	16192T; 16223T; 16298C; 16325C; 16327T	73G; 249d; 263G; 290d; 291d; 309.1C; 315.1C	1	C1
LPAZ047	16129A; 16223T; 16298C; 16325C; 16327T	73G; 249d; 263G; 290d; 291d; 309.1C; 315.1C	1	C1
LPAZ048	16111T; 16223T; 16290T; 16319A; 16362C	73G; 146C; 153G; 263G; 309.1C; 309.2C; 315.1C[a]	1	A2
LPAZ049	16183C; 16189C; 16217C; 16319A; 16360T[a]	73G; 146C; 150T; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ050	16182C; 16183C; 16189C; 16217C; 16293G; 16311C[a]	73G; 143A; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4c1a
LPAZ051	16183C; 16189C; 16217C; 16362C[a]	73G; 103A; 146C; 151T; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ052	16223T; 16298C; 16325C; 16327T	73G; 79T; 249d; 263G; 290d; 291d; 309.1C; 315.1C[a]	1	C1
LPAZ053	16176T; 16183C; 16189C; 16217C; 16240G; 16289G; 16362C[a]	73G; 204C; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ054	16092C; 16145A; 16223T; 16298C; 16325C	73G; 185A; 249d; 263G; 290d; 291d; 315.1C	1	CZ
LPAZ055	16051G; 16129A; 16182C; 16183C; 16189C; 16217C; 16234A[a]	73G; 152C; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ056	16092C; 16183C; 16189C; 16217C; 16362C[a]	73G; 143A; 146C; 215G; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ057	16182C; 16183C; 16189C; 16217C; 16362C[a]	73G; 146C; 215G; 263G; 309.1C; 309.2C; 315.1C[a]	1	B4
LPAZ058	16223T; 16293G; 16325C; 16362C	97A; 106d; 107d; 108d; 109d; 110d; 111d; 263G; 315.1C	1	D4j
LPAZ059	16223T; 16290T; 16319A; 16362C	73G; 146C; 153G; 182T; 194T; 195C; 235G; 263G; 315.1C	1	A4
LPAZ060	16223T; 16298C; 16311C; 16325C; 16327T	73G; 150T; 249d; 263G; 290d; 291d; 309.1C; 315.1C	1	C1
LPAZ061	16223T; 16298C; 16325C; 16327T	73G; 162Y; 249d; 263G; 290d; 291d; 309.1C; 309.2C; 315.1C[a]	1	C1
LPAZ062	16172C; 16223T; 16256T; 16298C; 16325C; 16327T	73G; 249d; 263G; 290d; 291d; 309.1C; 315.1C	1	C1
LPAZ063	16111T; 16217C; 16223T; 16290T; 16319A; 16343T; 16362C	73G; 146C; 235G; 260A; 263G; 315.1C	1	A2
LPAZ064	16223T; 16298C; 16325C; 16327T; 16345T	73G; 146C; 249d; 263G; 290d; 291d; 309.1C; 315.1C	1	C1
LPAZ065	16223T; 16298C; 16325C; 16327T; 16381C	73G; 152C; 249d; 263G; 290d; 291d; 309.1C; 315.1C[a]	1	C1
LPAZ066	16037G; 16223T; 16298C; 16325C; 16327T	73G; 222T; 249d; 263G; 290d; 291d; 309.1C; 315.1C	1	C1
LPAZ067	16183C; 16189C; 16217C; 16270T; 16278T[a]	73G; 94A; 152C; 183G; 204C; 263G; 309.1C; 315.1C[a]	1	B4
LPAZ068	16093C; 16183C; 16189C; 16223T; 16298C; 16325C; 16327T; 16381C[a]	73G; 249d; 263G; 290d; 291d; 315.1C	1	C1
LPAZ069	16111T; 16217C; 16223T; 16290T; 16319A; 16343T; 16362C	73G; 146C; 153G; 235G; 260A; 263G; 315.1C	1	A2
LPAZ070	16183C; 16188T; 16189C; 16217C	73G; 106d; 107d; 108d; 109d; 110d; 111d; 186T; 263G; 309.1C; 315.1C	1	B4
LPAZ071	16092C; 16111T; 16223T; 16290T; 16319A; 16362C	73G; 146C; 153G; 222T; 235G; 263G; 309.1C; 315.1C	1	A2
LPAZ072	16140C; 16223T; 16298C; 16311C; 16325C; 16327T	73G; 249d; 263G; 290d; 291d; 309.1C; 309.2C; 315.1C[a]	1	C1