B155 Mitochondrial DNA Population Data From El Salvador (Central America)

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After attending this presentation, attendees will learn the mitochondrial DNA data from 142 unrelated mestizo individuals from El Salvador, and their utility for human identification and population or anthropological studies.

This presentation will impact the forensic community and/or humanity by demonstrating the importance of specific data from different populations in order to perform more exact statistical calculations in forensic science, paternity analysis, and human evolutionary studies.

Mitochondrial DNA has been studied extensively in order to describe human variation, population substructure, and infer information about human migration patterns. There are several characteristics of mitochondria that make them useful for these roles. Mitochondria are maternally inherited and their genome is believed to be free of recombination. Therefore, the mitochondrial genome can be treated as a single locus with an effective population size that is ¼ of the size for nuclear autosomal genes. MtDNA also has been observed to have high mutation rates which contribute to the high levels of variability. Within the mitochondrial DNA the hypervariable regions (HVI and HVII) are thought to evolve particularly rapidly. The HVI includes nucleotide positions 16024 to 16365. The HVII includes nucleotide positions 73 to 340.

The high variability in mtDNA sequence has been used by forensic scientists for identity testing in both criminal and missing person cases. When one cannot exclude that an evidence sample and a reference sample may originate from the same source based on mtDNA analysis, an inference of the rarity of the sequence is required. To estimate the frequency of a mtDNA profile, population databases are used in forensics.

In this study the population of El Salvador, Central America was used as the target of analyses, due to the fact that there are not previous studies on this type of genome for this country. The population of El Salvador is composed by a 90% of mestizo (Amerindian-Spanish), 1% Amerindian and 9% white people. Samples of 142 unrelated mestizo individuals living in San Salvador were collected. DNA from blood or saliva was analyzed to get sequences of HVI and HVII following previous international recommendations. The extraction method used was the FTA® cards (Whatman, Florham Park, NJ). DNA was purified in a 1.2 mm punch, according to the manufacturer recommendations, followed by amplification with the correct primers that covered the positions 16024-16365 for HVI and 73-340 for HVII. Sequencing was carried over in an ABI 310 (Applied Biosystems, Foster City, CA) using Big Dye terminator v1.1 kits.

This table shows the polymorphisms observed more than one time, as well as the haplogroup to which they belong.

mtDNA HV1 region	mtDNA HVII region	Times observed	Haplogroup
111 209 223 290 319 362	73 146 153 235 263 309+C 315+C	3	A
111 223 290 299 319 362	73 146 153 235 263 309+C 315+C	3	A
153 223 240 290 319 362	73 146 153 235 263 309+C 315+C	2	A
111 290 319 362 391	73 146 153 235 263 315+C	2	A
111 223 290 319 362	73 146 153 235 263 309+C 315+C	6	A
183 189 193+CC 223 259 298 325	73 195 249d 263 290d 291d 315+C	2	C
111 187 223 290 319 362	73 146 153 235 263 309+CC 315+C	2	A
111 223 290 319 362	73 146 150 235 263 315+C	2	A
183 189 223 259 298 325 327	73 195 249d 263 290d 291d 315+C	2	C
111 223 290 319 362	73 146 152 153 197 235 263 309+C 315+C	2	A
223 290 311 319 362	73 146 153 235 263 309+C 315+C	2	A
111 187 223 290 319 362	73 146 153 235 263 309+C 315+C	2	A
111 223 290 319 362 391	73 146 153 235 263 309+C 315+C	4	A
111 172 223 256 290 319 362	73 146 153 195 235 263 309+CC 315+C	2	A
111 223 290 319 362	73 146 153 235 263 309 309.2C 315.1C	2	A
223 290 319 362	73 146 153 235 263G 315.1C	2	A
75 111 223 290 319	73 146 153 235 263 309.1C 315.1C	2	A

Out of the 142 samples there are 100 unique haplotypes; there are 13 haplotypes found twice (1.41%), two haplotypes found three times (2,11%), one haplotype found four (2,82%) times and another one found six times (4,22%). Most of the samples belong to haplogroup A, haplogroup C being the second most frequently observed group.

These data show enough variability of the population from El Salvador for the mitochondrial DNA hypervariable regions one and two, and hence they support the use of mtDNA analysis for identification purposes.

Mitochondrial DNA, Forensic Science, El Salvador