IDENTIFICATION OF HUMAN REMAINS RESCUED FROM OCEAN WATER BY MITOCHONDRIAL DNA ANALYSIS - DETECTION OF 25 POLYMORPHISMS

A.C.S. Góes[1], D.A Silva[1], J.C. Alvarez[2], J.A. Lorente[2] and E.F. Carvalho[1]
[1]DNA Diagnostic Laboratory, Rio de Janeiro State University, Rio de Janeiro, Brazil
[2]Laboratory of Genetic Identification, University of Granada, Granada, Spain[2]

Skeletal human remains found submersed under water in the coast of Rio de Janeiro State, Brazil, in an advanced state of decomposition were brought to the Legal Medicine Institute to be identified. The human remains were rescued from the ocean water more than 3 months after the death report. Bone mitochondrial DNA was amplified at HVI and HVII regions and sequenced. From the total of 25 polymorphisms, 11 were found at HVI and 14 were found at HVII. Transition C to T or T to C was the predominate polymorphism at HVI. In 11 polymorphisms, only one transition A to G was detected. Transversions were not observed. For HVII, transitions were also the most common polymorphism, but transition C to T or T to C is equal to A to G or G to A polymorphism. In 14 polymorphisms, we detected 5 of each one. Furthermore, we found 2 transversions, one A to C (position186) and another C to A (position 189) in HVII. Studies of mitochondrial DNA population data showed that African, African American and Hispanic samples demonstrated a large portion of their C to A and A to C transversions in the HVII region at sites 186 and/or 189. The 2 insertions occurred in the HVII region at sites 309.1 and 315.1, within the stretch of C. The 25 variations proved to be typical of the African L1c haplogroup, indicating the African ancestry of the white woman. In a study concerning Brazilian mtDNA lineages, the analysis of 247 white individuals from the five major geographic regions of the country have shown a contribution of 28% from the African group to the total mtDNA pool. The L1c subgroup contributes 19% of total African group. Sequence analysis of human mitochondrial DNA has been demonstrated to be a valid and reliable tool for the genetic characterization of forensic biological specimens. Typically, mtDNA analysis is attempted on samples for which nuclear DNA typing is not likely to be successful. In this case, only this kind of analysis would be applied as the skeletal human remains found submersed under salted water didn't yield DNA suitable for STR nucleus *loci* amplification. We were able to correlate the mitochondrial DNA of a claimed mother and the human remains that were supposed to be from her son who disappeared months ago. To convey the rarity of a mtDNA type among unrelated individuals, the current practice is based on the number of times a particular sequence is observed in a database. As this haplotype was not related in a database of 2427 individuals, the identification is directly done.

Granted by Foundation for Research Support in Rio de Janeiro (Faperj) and M. Botin Foundation.