Mitochondrial DNA: Sequence Variations in Argentine Population

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A total of 380bp of the non-coding region (D-Loop), Segment I of human mitochondrial DNA (mtDNA) from 107 unrelated Argentine individuals have been determined. One hundred two blood samples and five bone samples of a forensic origin were analyzed. At the B.N.D.G. mtDNA bone studies constitute a much greater number than we present here, but due to DNA degradation, the number of nucleotides was low, for this reason they were excluded from the present communication. Comparison of these sequences with the previously determined for the human mitochondiral genome has revealed 80 changed sites (21.05%), which, in all cases, were nucleotide substitutions. At three sites, two different kinds of nucleotide substitutions were observed. Fifty percent of mutations in the Argentine population were a single event, while the nucleotide change in position 16223 (C-T), was found in other populations groups. In the whole data, transition types of substitution were more prominent than transversions; that is, 95% were transitions while only 5% were transversions. Moreover, transitions between pyrimidines were more prevalent than those between purines. None of the mutations in those samples were a marker for Argentine population. A computer software was designed to compare differences per pairs of individuals in nucleotide substitutions.

These data are important for the recognition of the variability of nucleotides in the human mitochondrial genome, in the samples randomly studied in Argentine population. This type of study currently represents the only tool suitable for determining familiar relationships when only one relative is available.