Amerindian mitochondrial DNAs have rare Asian mutations at high frequencies, suggesting they derived from four primary maternal lineages

ABSTRACT

The mitochondrial DNA (mtDNA) sequence variation of the South American Ticuna, the Central American Maya, and the North American Pima was analyzed by restriction-endonuclease digestion and oligonucleotide hybridization. The analysis revealed that Amerindian populations have high frequencies of mtDNAs containing the rare Asian RFLP HincII morph 6, a rare HaeIII site gain, and a unique AluI site gain. In addition, the Asian-specific deletion between the cytochrome c oxidase subunit II (COII) and tRNA(Lys) genes was also prevalent in both the Pima and the Maya. These data suggest that Amerindian mtDNAs derived from at least four primary maternal lineages, that new tribal-specific variants accumulated as these mtDNAs became distributed throughout the Americas, and that some genetic variation may have been lost when the progenitors of the Ticuna separated from the North and Central American populations.

Keyword: Mitochondrial DNA (mtDNA); Amerindian; Gene frequency; Mutation; Asia