POLYMORPHISM IN MITOCHONDRIAL DNA SEQUENCE ANALYSIS OF FORENSIC CASES

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Polymorphism and maternal inheritance of mitochondrial DNA control regions are two critical factors in their application in evolution, human population studies and forensic case analysis. Although the basis for polymorphism relates to homoplasmic and heteroplasmic mutations, little is known about their actual contribution in forensic cases. Therefore, the following study was undertaken to examine the contribution for both mutations to polymorphism in well-characterized different forensic cases. The biological source for these forensic samples included hair roots, hair shafts, skeletal tissues and various other tissues. All forensic cases showed a mutational rate of 10% in mtDNA control region. However, the mutational rate at HV-I (12%) was relatively higher than that observed at HV-II (9%). In a total of 66 homoplasmic mutations, 57 transitions, one transversion, three deletions and five insertions were found in a number of cytosine residues. Two additional novel homoplasmic mutations were also identified. One of them is a transitional mutation (T>C) at nucleotide number 16117 and the other is a deletion mutation at nucleotide number 291 (A-del). In seven specimens, 22 heteroplasmic mutations were identified and their number ranged between one to seven nucleotides per specimen.

Our data analysis indicated that polymorphism in mitochondrial DNA sequence analysis is a powerful tool in human identification.