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Deciphering the spectrum of somatic mutations in the entire mitochondrial DNA genome

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ABSTRACT. The mitochondrion is a crucial intracellular organelle responsible for regulating cellular energy metabolism, producing free radicals, initiating and executing the apoptotic pathways. Previous studies have shown that somatic mutations in mitochondrial DNA are associated with various tumors, which may be involved during carcinogenesis and tumor progression. To examine the mutation pattern in cancer, 625 reported somatic mutations in the mitochondrial DNA genome were analyzed. We found that, except for deletions and insertions, most somatic mutations were point mutations, accounting for 89.44% of somatic mutations. Transition was the predominant form of

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somatic mutation in the entire mitochondrial DNA genome, accounting for 87.12% of point mutations, most of which were homoplastic. Frequency statistics analysis of point mutations indicated that, except for 3 tRNA genes, the mutations were distributed on all resting genes and in the D-loop region, with the latter showing the highest frequency of somatic mutation (19.34%), followed by the tRNA leucine 2 gene and non-coding regions between base pairs 5892 and 5903, while 13 coding-region genes and 2 rRNA genes showed a relatively lower frequency of somatic point mutations. Nonsynonymous mutations and terminal amino acid changes were the primary point somatic mutations detected from 13 coding-region genes, which may cause mitochondrial dysfunction in cancer cells. We found that the somatic mutations may affect the mitochondrial DNA genome; the non-coding region should be examined to identify somatic mutations as potential diagnostic biomarkers for early detection of cancer.

Key words: Cancer; Mitochondrial DNA genome; Somatic mutation

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