Tandem duplication/triplication correlated with poly-cytosine stretch variation in human mitochondrial DNA D-loop region

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摘要

Abstract

Somatic mutations in the mitochondrial DNA (mtDNA) displacement loop (D-loop) region have been frequently detected in various human cancers. In a previous study, we identified a polyplasmic 260-bp tandem duplication and triplication mutation in the mtDNA D-loop of one gastric cancer. In the present study, we adopted a more sensitive back-to-back polymerase chain reaction method to screen for this 260-bp tandem duplication/triplication in 197 cancers and their adjacent non-cancerous tissues. Nine samples of primary cancer (4.6%) were found to harbor the tandem duplication/triplication and these were made up of four out of 31 (12.9%) gastric cancers, two out of 45 (4.4%) breast cancers, two out of 56 (3.6%) hepatocellular cancers and one out of 32 (3.1%) colon cancers, but no tandem duplication/triplication was present in any of 33 lung cancers. We also found an expanded and polyplasmic poly-cytosine (poly-C) stretch around nucleotide position (np) 568 in eight of the 197 (4.1%) cancer patients. All the eight cancer samples carried the 260-bp tandem duplication/triplication. In addition, we detected the np 568 poly-C length variations in 11 of 234 (4.7%) peripheral blood samples of non-cancer population and the 260-bp tandem duplication in nine of the 11 cases with the np 568 poly-C length variations. These observations suggest that the occurrence of the tandem duplication/triplication in mtDNA D-loop is not specific for cancer tissues, but highly associated with the poly-C length variations around np 568.