A Novel SNP Genotyping Method Using Multi-Color Fluorescence Labeled Ligase Detection Reaction and High Resolution CE-SSCP

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For the development of clinically useful genotyping methods for single-nucleotide polymorphisms (SNPs), accuracy, simplicity, sensitivity, and cost-effectiveness are the most important criteria. Among the methods developed SNP genotyping technology, the ligation-dependent method is considered the simplest for clinical diagnosis. However, sensitivity is not guaranteed by itself, and analysis of multiple targets is limited by the detection method. Although capillary electrophoresis (CE) is an attractive method to detect multiple targets, the multiplex assay process is complicated because of the size-based DNA separation principle. In this study, we employed the ligase detection reaction (LDR) coupled with high-resolution CE-based single-strand conformation polymorphism (CE-SSCP) to develop robust SNP genotyping method. Simple and sensitive SNP analysis can be performed using this method involving the use of similar-sized probes, without complex probe design steps. We found that it could not only accurately discriminate base mismatches but quantitatively detect 37 SNPs of the tp53 gene, which are used as targets in multiplex analysis, using three-color fluorescence-labeled probes.