A multiplex and sensitive SNP genotyping assay using multi-color fluorescence labeled ligase detection reaction-based mismatch discrimination and CE-SSCP

최 웅, 정규열†
포항공대
(gyjung@postech.ac.kr†)

Accuracy, simplicity, sensitivity and cost-effectiveness are the most important criteria for a genotyping method for single-nucleotide polymorphisms (SNP). One method developed for SNP genotyping, ligase-based coupled capillary electrophoresis (CE) is considered for clinical diagnosis. However, conventional CE system is the design process and multiplex assay procedure are complicated because of the DNA size-based separation principle. In this study, we developed a simple, accurate and sensitive multiplex genotyping method using ligase detection reaction and high-resolution CE-based single-strand conformation polymorphism (CE-SSCP). With this high-resolution CE-SSCP system, we were able to use similar-sized probes, simplifying the design step and assay process. We found that this method could accurately discriminate single base mismatches on SNPs of tp53 gene used as targets for multiplex detection using multi-color fluorescence labeled probes.