Diagnosis of *BRCA1* mutations using SBE method with zip-code microarray

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We described multiplex assay to detect Korean-specific breast cancer susceptibility genes (*BRCA1*)[1] using single base extension (SBE) method with zip-code microarray[2]. we performed multiplex SBE reactions for the diagnosis of seven mutations in *BRCA1* with the wild-type, and heterozygote mutant samples in a blinded manner. These seven mutations were divided into 4 different PCR reactions. The four PCR amplification products were pooled in order to be used as template for the subsequent multiplex SBE reaction. We added 9 chimeric primers corresponding to the 9 zip-code microarrays to a single tube and performed four different multiplex SBE reactions with each sample, one for each biotin-labeled ddNTP. The result shows high fidelity genotype discrimination for seven mutations in one-step single base extension reaction. With the wild-type homozygote sample, the signals were generated only at the zip positions corresponding to the homozygote allele. When we tested the mutant sample with four heterozygote alleles, two signals were correctly generated at each of the four zip positions corresponding to the heterozygote alleles. These results demonstrated the strategy developed in this work using four separate SBE reactions with zip-code microarray could be used for the diagnosis of *BRCA1* mutations

References

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