Diagnosis of human genetic mutations based on DNA microarray technology

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Abstract

In this presentation, we will discuss several recent achievements developed in my laboratory for microarray-based diagnosis of human genetic mutations including HNF-1 and BRCA1 mutations. To determine the presence of the genetic mutations in a human sample, we prepared allele-specific oligonucleotide chips from selected mutation sites and generated target probes using a two-step method for Cy-3 DNA samples[1] or *in vitro* transcription of promoter-tagged PCR products for Cy-3 RNA samples[2]. Hybridization of the target probes to the chips successfully identified all of the genotypes for the tested sites. For more reliable diagnosis, we also employed single base extension (SBE) reaction and zip-code microarray technique for our strategy. Particularly we developed an efficient PNA zip-code microarray for the detection of HNF-1α mutations[3]. Using multiplex SBE reactions and zip-code strategy, we were able to correctly diagnose several mutation sites in exon 2 of HNF-1α with a wild-type and mutant samples including a MODY3 patient. These works represent successful applications of DNA microarray technology for the diagnosis of human genetic mutations.

References

1. Seong-Chun Yim, Hyun Gyu Park, Ho Nam Chang, and Dae-Yeon Cho, "Array based mutation detection of BRACA1 using direct probe/target hybridization" (2005), Analytical Biochemistry, 337, 332-337.
2. Hyun Gyu Park, Hyun Ok Ham, Kyung Hee Kim, and Nam Huh, "Oligonucleotide chip for the diagnosis of HNF-1 mutations" (2005) Biosensors and Bioelectronics, in press.
3. Jae Yang Song, Hyun Gyu Park, Sung-Ouk Jung, and Jae Chan Park, "Diagnosis of HNF-1 α mutations on a PNA zip-code microarray by single base extension" (2005), Nucleic Acids Research, 33 (2), e19.