

研究简报

连接反应介导的等位基因特异性扩增-微流控芯片电泳法同时检测多个SNP位点

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摘要 以CYP2D6基因中的6个SNP位点为测定对象, 开展多个SNP位点同时测定的方法学研究.

关键词 [ALM-ASA](#) [微流控芯片电泳](#) [SNP](#) [CYP2D6](#)

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Simultaneous Detection of Multiplex SNPs by Adapter-ligation Mediated Allele-specific Amplification(ALM\|ASA) on CE-chip Electrophoresis Device

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Abstract To detect multiplex single nucleotide polymorphisms(SNPs) simultaneously, a new method was established by combining ALM-ASA with microfabricated CE-chip. Taking the CYP2D6 gene as an example, six SNPs, 100C>T(P34S), 1707T>del(frameshift), 1758G>T(stop codon), 2470T>C, 2549A>del(frameshift) and 2613AGA>del(K281del), were typed by four steps consisting of preamplification, digestion and ligation, allele-specific amplification, and amplicon separation by chip-CE. The genotyping results of 20 different genome samples by 6-plexed ALM-ASA were completely consistent with those obtained by polymerase chain reaction-restriction fragment length polymorphism analysis(PCR-RFLP), indicating that the multiplex approach established in this study was accurate and inexpensive. As the small reagent consumption by CE-chip device, a low cost for SNP typing was achieved together with the multiplex PCR technology proposed in this report. Neither modification of microchip channels nor clean-up process of PCR products was required; this greatly shortens the whole time for SNP typing.

Key words [ALM-ASA](#) [Microchip electrophoresis](#) [SNP](#) [CYP2D6](#)

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