

研究报告

# 单核苷酸多态性检测方法的研究进展

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**摘要** 单核苷酸多态性 (single nucleotide polymorphism, SNP) 的研究已成为人类后基因组时代的主要内容之一。因此建立高度自动化和高通量的SNP检测分析技术十分重要。文章系统地介绍了最新发展的几种SNP检测技术的原理和检测平台, 详细阐述了等位基因特异性杂交、内切酶酶切技术、引物延伸法、寡核苷酸连接反应等SNP检测原理, 以及平板读数仪、基因芯片、微球阵列技术和质谱仪等检测平台, 并对SNP高通量检测技术的发展进行了展望。

**关键词** [单核苷酸多态性](#); [检测技术](#); [检测平台](#)

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## Approaches for SNP Genotyping

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### Abstract

With the completion of the Human Genome Project (HGP), typing single nucleotide polymorphisms (SNP) has become one of the main tasks in the post-genome era. Consequently, a robust, flexible and cost-effective technique for SNP typing is essential to analyze a large number of SNPs. The latest genotyping technologies and the relative detection platforms were introduced systematically. The principle of SNP typing was described in detail in respect of allele-specific hybridization, restriction enzyme digestion, primer extension and oligonucleotide ligation assay, as well as the genotyping platforms of plate readers, genechips, bead array and mass spectrometry. Moreover, the way to the high-throughput genotyping in the future was briefly discussed.

**Key words** [SNP](#) [genotyping technologies](#) [genotyping platforms](#)

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