

单核苷酸多态性检测方法的新进展 Progress in Detection Methods of Single Nucleotide Polymorphisms

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摘要

单核苷酸多态性 (single nucleotide polymorphism, SNP) 是第三代遗传标记, 在基因定位、遗传疾病和人类起源等理论研究中具有重大意义, 在抗药性或药物过敏反应中扮演着极其重要的角色, 正逐步成为分子诊断、临床检验、新药研发的重要手段。随着人类基因组测序的完成, SNP分型和发现成为遗传和生命医学领域研究的热点之一。近年来, SNP的检测方法层出不穷, 发展很快。文章综述和分析了几种新建立的SNP检测方法, 包括基因芯片、分子探针、荧光偏振、荧光共振、质谱和磁性颗粒分析。在生物化学、工程学和软件分析等方面取得突破的基础上, 有望建立灵敏准确、简便易行、高通量、低费用的SNP技术。Abstract: Single nucleotide polymorphism (SNP) is the third generation genetic marker. SNP detection now is becoming increasingly important means in molecular diagnostics, clinical assay and novel drug development. It plays an essential role in drug resistance and anaphylactic reaction and has the importance in theoretical studies of gene location, hereditary diseases and human origin. With the accomplishment of human genome sequencing, the genotyping and discovering of SNP are becoming hot subjects in genetics and biomedicine researches. The methods for SNP detection were renewed rapidly and developed fast in past few years. In this review, several newly established detection methods including gene chip, molecular probe, fluorescence polarization and resonance, mass spectrometry, and bacterial magnetic particle are discussed. It could be expected that an accurate and sensitive, simple and easy-to-handle SNP technology with low cost and high throughput will be available on the basis of research breakthroughs of biochemistry, engineering and analytic software.

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Abstract

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