分子生物学技术在遗传病诊断中的应用 The Application of Molecular Biological Techniques in Diagnosis of Human Genetic Diseases

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摘要 论述了遗传性疾病的发病机理和遗传病诊断的途径和策略,着重介绍了人类遗传疾病基因诊断技术的原理和发展状况,对该领域的新成果作了介绍和总结,对发展动向和存在的问题作了简要论述。

Abstract: The mechanisms of human genetic diseases and strategies to test human genetic diseases are discussed in the review. The development of the gene diagnosis techniques to genetic diseases and their principles are introduced emphatically. The new achievements in the field are valued and concluded, and the prospect and the difficulties in genetic diseases diagnosis are also briefly discussed.

关键词 遗传疾病 基因诊断 分子生物学技术 Key words genetic disease gene diagnosis molecular biological technique

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