

1. Record Nr.	UNINA9910791285803321
Titolo	Molecular diagnostics [[electronic resource] /] / edited by George Patrinos, Wilhelm Ansorge
Pubbl/distr/stampa	London, : Academic, 2010
ISBN	1-282-73741-4 9786612737411 0-08-092318-6
Edizione	[2nd ed.]
Descrizione fisica	1 online resource (617 p.)
Altri autori (Persone)	PatrinosGeorge P AnsorgeWilhelm <1944->
Disciplina	616.0756
Soggetti	Molecular diagnosis Genetic screening
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Front Cover; Molecular Diagnostics; Copyright Page; Contents; List of Contributors; Preface - First Edition; Preface - Second Edition; Foreword - First Edition; Chapter 1. Molecular Diagnostics: Past, Present, and Future; 1.1 INTRODUCTION; 1.2 HISTORY OF MOLECULAR DIAGNOSTICS: INVENTING THE WHEEL; 1.3 THE PCR REVOLUTION: GETTING MORE OUT OF LESS; 1.4 MOLECULAR DIAGNOSTICS IN THE POST-GENOMIC ERA; 1.5 FUTURE PERSPECTIVES: WHAT LIES BEYOND; 1.6 CONCLUSIONS; REFERENCES; Section I. Molecular Diagnostic Technology; Chapter 2. Allele-Specific Mutation Detection; 2.1 INTRODUCTION; 2.2 PCR-ARMS 2.3 PCR-ASO2.4 THE COMPETITIVE OLIGOPRIMING ASSAY; 2.5 CONCLUSIONS; REFERENCES; Chapter 3. Enzymatic and Chemical Cleavage Methods to Identify Genetic Variation; 3.1 INTRODUCTION; 3.2 CHEMICAL PROPERTIES OF MISMATCHES; 3.3 CHEMICAL CLEAVAGE OF MISMATCH METHOD FOR MUTATION DETECTION; 3.4 ADVANTAGES AND LIMITATIONS; 3.5 ENZYMATIC CLEAVAGE OF MISMATCH METHODS; 3.6 CONCLUSIONS; REFERENCES; Chapter 4. Mutation Detection by Single Strand Conformation Polymorphism and Heteroduplex Analysis; 4.1 INTRODUCTION; 4.2 PRINCIPLES OF SINGLE

## STRAND CONFORMATION POLYMORPHISM ANALYSIS

4.3 FLUORESCENT SINGLE STRAND CONFORMATION POLYMORPHISM ANALYSIS; 4.4 PARAMETERS INFLUENCING SINGLE STRAND CONFORMATION POLYMORPHISM ANALYSIS; 4.5 HETERODUPLEX ANALYSIS FOR MUTATION DETECTION; 4.6 SENSITIVITY AND LIMITATIONS; 4.7 DETECTION OF THE UNDERLYING GENOMIC VARIATION USING SSCP AND HDA; 4.8 CONCLUSIONS AND FUTURE ASPECTS; REFERENCES; Chapter 5. Capillary Electrophoresis; 5.1 INTRODUCTION; 5.2 HISTORY, PRINCIPLE, AND POTENTIAL APPLICATIONS OF CAPILLARY ELECTROPHORESIS; 5.3 CAPILLARY ELECTROPHORESIS IN MOLECULAR DIAGNOSTICS; 5.4 MODES OF APPLICATION; 5.5 SPECIFIC DIAGNOSTIC APPLICATIONS; 5.6 FUTURE IMPROVEMENTS; REFERENCES; Chapter 6. Temperature and Denaturing Gradient Gel Electrophoresis; 6.1 INTRODUCTION; 6.2 THE THEORY OF TEMPERATURE-GRADIENT GEL ELECTROPHORESIS; 6.3 THE PRACTICE OF TEMPERATURE GRADIENT GEL ELECTROPHORESIS; 6.4 DENATURING GRADIENT GEL ELECTROPHORESIS (DGGE); 6.5 THE USE OF TGGE/DGGE FOR MUTATION DETECTION; 6.6 DETECTION RATE AND SENSITIVITY; 6.7 RELATED TECHNIQUES AND VARIANTS; 6.8 TECHNICAL EQUIPMENT FOR TGGE/DGGE; 6.9 APPLICATIONS OF TGGE/DGGE AND RELATED METHODS; 6.10 CONCLUSIONS; ACKNOWLEDGEMENTS; REFERENCES; Chapter 7. Real-Time Polymerase Chain Reaction; 7.1 HISTORY OF PCR; 7.2 PRINCIPLE OF REAL-TIME PCR; 7.3 REAL-TIME THERMAL CYCLERS; 7.4 HOW DATA ARE OBTAINED; 7.5 HOW DATA ARE QUANTIFIED; 7.6 MULTIPLEX REAL-TIME PCR; 7.7 APPLICATIONS IN MOLECULAR DIAGNOSTICS; 7.8 CRITERIA FOR DEVELOPING REAL-TIME PCR ASSAYS; 7.9 CONCLUSIONS; REFERENCES; Chapter 8. Pyrosequencing; 8.1 INTRODUCTION; 8.2 TECHNOLOGY; 8.3 APPLICATIONS; 8.4 CONCLUSIONS; ACKNOWLEDGEMENTS; REFERENCES; Chapter 9. Application of Padlock and Selector Probes in Molecular Medicine; 9.1 INTRODUCTION; 9.2 PADLOCK AND SELECTOR PROBES; 9.3 APPLICATION OF PADLOCK AND MOLECULAR INVERSION PROBES FOR GENOTYPING

---

### Sommario/riassunto

**KEY FEATURES:** Contains state-of-the-art techniques for the detection of the underlying genetic heterogeneity leading to inherited disorders  
Includes in-depth discussion of ethical and safety considerations  
Identifies genetically modified organisms (GMOs)  
Covers forensic analysis and everyday issues in a diagnostic laboratory  
**DESCRIPTION:** The 2e of Molecular Diagnostics, the only book dealing with diagnosis on a molecular level, discusses current molecular biological techniques used to identify the underlying molecular defects in inherited diseases

---