

Preface

Diagnostic molecular biology is the integration of the knowledge and technology in molecular biology with clinical laboratory techniques. It is a clinical reality with its roots deep inside the fundamental understanding of genome structure and cellular activities at the molecular level. In the past few years, the field of molecular diagnosis has grown rapidly due to the revolutionized developments and discoveries in the fields of molecular genetics and biotechnology, and the need to provide diagnosis at the molecular level for all kinds of diseases. As the study of diagnostic molecular biology has increasingly gained attention, more and more clinical laboratories are preparing to deal with its demands and challenges, and molecular diagnosis has now become an important part of healthcare. As such, this book aims to deal with all aspects of diagnostic molecular biology from its principles and techniques to application of molecular diagnostics in a variety of fields.

This textbook is aimed at advanced students, entry-level scientists, and technicians who intend to enter the clinical laboratory professions with knowledge in general biology, genetics, and microbiology. In order to provide the most current understanding of the rapidly improving techniques in molecular diagnosis, this book is organized into three parts. Part 1 provides the fundamentals of molecular biology and comprises Chapters 1 through 5. Chapter 1 serves as the introduction to the structure and function of nucleic acids. Chapters 2–4 contain basic coverage of nucleic acid-based cellular activities including replication, repair, and gene expression. Chapter 5 discusses genome structure and evolution.

Part 2 describes commonly used molecular biology techniques and newly developed techniques in molecular diagnosis, including real-time PCR and NGS. It comprises Chapters 6 through 12. Chapters 6 to 8 cover the techniques commonly used in nucleic acid and protein preparation, analysis, and quantitation. Chapter 9 discusses amplification techniques for nucleic acids. Chapter 10 contains the techniques to characterize nucleic acids and proteins. Chapters 11 and 12 provide detailed information on the principles and applications of sequencing techniques.

Part 3 includes Chapters 13 through 16 and contains current molecular diagnostic techniques in identifying diseases. Chapter 13 covers molecular cytogenetic diagnosis in identifying chromosomal disorders. Chapters 14 and 15 discuss molecular diagnosis in genetic diseases and infectious diseases. Finally, Chapter 16 provides guidance for operating a molecular clinical laboratory.

This book gives an overview of important topics of interest within molecular diagnosis and shows the remarkable recent progress of molecular techniques in clinical analysis. The length is intended to provide instructors with a choice of favorite themes without sacrificing the important topics and without providing an overwhelming curriculum for the limited amount of time available in one semester. As such, this book can lead students to understand the essentials of molecular biology and to know how molecular techniques are used in clinical diagnosis.