Molecular Diagnostics
Molecular Diagnostics

For the Clinical Laboratorian

Second Edition

Edited by

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Foreword by

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Humana Press
First and foremost, we would like to dedicate this book to our wives, Monty and Nancy, for their unprecedented support of our careers and families.

To all of our colleagues, students, trainees, and mentors over the many years, we thank you for your continued support.
FOREWORD

In 1994 I coedited a book, Molecular Pathology: Approaches to Diagnosing Human Disease in the Clinical Laboratory, that coined the phrase “molecular pathology” to refer to applications of molecular biology in the traditional areas of laboratory medicine. That compilation of clinical molecular techniques included 11 chapters and an epilogue on “New Directions for the Clinical Laboratory.” Chapter headings included: automation, neoplasia, heritable diseases, and infectious diseases.

Three years later, in 1997, Bill Coleman and Greg Tsongalis edited Molecular Diagnostics: For the Clinical Laboratory, which contained 18 chapters and introduced the new areas of RT-PCR and In Situ PCR, and included a section on gene therapy. Now, ten years after the introduction of molecular pathology, we are looking at the second edition of Bill Coleman and Greg Tsongalis’ text.

This second edition now comprises 44 chapters, and contains new chapters on bioinformatics, microarrays, methylation assays, FISH, laser capture microdissection, quality assurance, chimerism studies for bone marrow transplantation, and separate chapters on genetic counseling and ELSI (ethical, legal, and social issues). These new chapters represent developments in the past decade that have fundamentally changed the scope from molecular pathology to molecular diagnostics, highlighting the changing role of the clinical laboratory who direct these efforts. It should be evident that molecular pathology and diagnostics impact almost every conceivable subspecialty in laboratory medicine and, particularly in the case of pharmacogenomics, have led to the development of new areas of investigation.

What is less evident is the burden placed upon the laboratorians directing these efforts. Issues regarding training, certification, continuing education, and reimbursement (just to name a few) have demanded enormous time and effort from professional organizations and governmental agencies. In addition, because conventional approaches cannot always be applied to molecular diagnostics, unique solutions for quality assurance and quality control must be developed. These challenges have engendered committees, subcommittees, taskforces, and workgroups from regulatory agencies and professional organizations, representing worldwide constituencies. As in any situation involving so many players, consensus-building and communication are mandatory. To these ends, professional journals and textbooks are our best hope for remaining current with this rapidly changing field. One needs only to compare the wealth of knowledge in this current edition with our efforts of only ten years ago to appreciate the magnitude of this challenge.

Lawrence M. Silverman, PhD
It has been almost ten years since the concept for producing the first edition of *Molecular Diagnostics: For the Clinical Laboratorian* was conceived. In those ten years the field of molecular pathology and diagnostics has exploded as many predicted. The clinical diagnostic laboratory continues to function as the playing field for this expansion that includes vast and dynamic changes in test menus, instrumentation, and clinical applications. The impact of this field on the routine practice of clinical medicine and management of patients continues to be felt as new developments that span all areas of laboratory medicine exceed our expectations.

The success of this technology in a clinical setting is highly dependent upon the training of well-qualified technologists, residents, and clinicians alike, who will not only have to perform and interpret results of these tests but also understand the limitations of the technology and resulting clinical implications. The production of this second edition is a testament to our passion and commitment for the teaching and training of qualified individuals who wish to embark on this journey. The numerous training programs, educational venues, and board certification examinations that have evolved over the past ten years also send a strong vow of commitment by others in the field to ensure the successful use of these new tools in supporting the best possible patient care that is available.

The second edition of *Molecular Diagnostics: For the Clinical Laboratorian* begins with a historical perspective of laboratory medicine followed by an overview of basic molecular biology techniques and concepts. Part III provides a more in depth examination of some advanced molecular technologies and their potential uses. Part IV describes other technologies found in the clinical laboratory that can complement or be complemented by molecular diagnostic technologies. The increasing need for awareness and practice of quality assurance in this field led us to include a complete section (Part V) that examines some of these issues. Although the first edition included clinical applications all in one section, the increased number of applications led us to develop separate sections for genetic disease, human cancers, infectious diseases, and identity testing (Parts VI–IX). Finally, the book concludes with a section on genetic counseling and ethical/social issues involved with nucleic acid testing.

Although no such book could possibly be all encompassing in such a rapidly developing field, we feel that the material covered herein will provide the reader with an excellent overview.

William B. Coleman  
Gregory J. Tsongalis
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