
This highly successful and comprehensive textbook of clinical chemistry has become one of the two authoritative texts in the field. The new edition has 57 chapters, 9 appendices, and 113 contributors (up from 96 in the 2nd edition). The author list is a who’s who of clinical chemistry today.

In a major organizational change, the 3rd edition includes analytical methods at the end of each pathophysiology chapter rather than in a separate section at the end of the book. This greatly improves the continuity of the presented topics. In addition, because most laboratories now use prepackaged reagents for routine chemistry, the editors have reduced the discussion of analytical methods, particularly those that are no longer in use. The 3rd edition is one of the few examples of a “restructuring” project that laboratorians can embrace.

Five new chapters have been added to the text: Laboratory Management, Mass Spectrometry, Point-of-Care (Near-Patient) Testing, Laboratory Evaluation of Transplant Recipients and Donors, and Addiction and Substance Abuse. Several other chapters have been substantially modified or updated, including Liquid Chromatography, Laboratory Information Systems, Immunochemical Techniques, Automation, Laboratory Statistics, and Reference Intervals and Clinical Decision Limits. Expanded versions of these chapters were warranted because dramatic changes have occurred in these fields in the seven years since publication of the last edition. A section on total laboratory automation (robotics) might also have been included, although this topic is more general to all of laboratory medicine.

My only criticism of this and other books of its genre is that it contains little information as to how clinicians and laboratorians can effectively use the many analytes described therein. Evidence-based clinical practice guidelines (CPGs) have been prepared on most of the diseases covered in this book, with recommendations as to what, when, and where tests should be ordered, and how results are used in patient management. In order to survive, practicing clinical chemists need information on outcomes studies involving laboratory tests, and must be able to assist in establishing CPGs at their own institutions. It is no longer enough for a clinical chemistry textbook to catalog all analytes, methodologies, and instrumentation.

In summary, this book is an absolute must for anyone who teaches clinical chemistry or laboratory science, or who is responsible for conducting testing in a hospital or reference laboratory. Individuals who work in the in vitro diagnostics industry will also want this book on their reference shelf to support their research and product development efforts. With regard to value, I am amazed that a reference textbook of over 1000 pages can be offered for under $70.00. That’s less than three Danielle Steele (hardback) novels!

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Molecular Diagnosis of Genetic Diseases stands nicely apart from a burgeoning list of similar textbooks on clinical molecular genetics. Although different authors contributed its 20 chapters, each chapter is similarly formatted and the book well edited, so it does not read as if it was written by a host of specialists. Moreover, in a disease-specific manner, the text addresses exactly how to perform molecular diagnostic testing, often delving into the nitty gritty of each assay, and encompasses techniques ranging from allele-specific oligonucleotide hybridization to protein truncation tests.

In the first chapter, the editor succinctly explains why family studies set such testing apart from routine molecular diagnostics, and what special constraints inheritance across generations places on the clinical laboratory relating to long-term specimens and information storage and retrieval. Five different categories of clinical molecular genetic testing are defined (differential diagnosis, carrier detection within families and within populations, prenatal diagnosis, and presymptomatic screening) along with frank discussions of other important issues such as laboratory staffing, training, quality assurance, accreditation, and even audits. Eight chapters deal with specific common molecular genetic diseases: muscular dystrophy, unstable trinucleotide repeats, familial adenomatous polyposis, cystic fibrosis, congenital adrenal hyperplasia, hemoglobinopathies, Prader Willi and Angelman syndromes, and analysis of X chromosome inactivation. Each chapter contains a brief introduction, strategies for analysis, methods and materials, and multiple examples, interpretations, notes, and references. Two chapters are devoted solely to genetic counseling and quality assurance. The remaining chapters largely concern technique and cover such topics as sequencing-based diagnosis, microtiter array diagonal and pulsed field gel electrophoresis, as well as high throughput single-strand conformation polymorphism analysis.

The book’s scope, crisp text, attention to details, and spiral binding all make this a useful benchtop reference manual, to which this already stained copy will testify.

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