Preface

Today, molecular and genomic information is informing the patient care decisions in many, if not most, areas of healthcare. Clearly, cancer diagnosis, prognosis, and treatment are driven largely by the molecular variants that drive the cancer and are the targets for new therapies. Medical genetics is moving beyond the classic single gene genetic disorders as we understand the genetic risk factors that drive the common chronic diseases that are costly to our healthcare system. While the clinical relevance of all areas of the human genome is not yet understood, our knowledge is growing rapidly and expanding well beyond the protein-coding genes to include many regulatory-coding regions, such as microRNAs and long noncoding RNAs (lncRNAs), in regions of the genome which used to be considered "junk." For infectious diseases, we are beginning to understand not only the well-known and emerging infectious agents, but that health and disease also relates to the symbiotic relationship of each patient with their microbiomes. Finally, the technologies available to the clinical molecular laboratory have advanced so the genome of individual patients can be analyzed for clinical care, even resulting in the definition of genomic critical values, which are recommended to be reported any time an exome or genome is sequenced for clinical purposes.

Molecular Pathology in Clinical Practice addresses all areas of clinical molecular pathology practice in a single textbook. This second edition has 12 new chapters, in addition to updates on the chapters from the first edition. The new chapters cover diseases not included in the first edition, plus two chapters on next-generation sequencing applications in genetics and cancer, and a proteomics chapter. The purpose of this textbook remains to provide a comprehensive reference for the practicing molecular pathologist as well as a resource for pathologists in any area of practice. The book also will continue to be used by training programs, both for Anatomic and Clinical Pathology and for Molecular Genetic Pathology trainees. This book is not meant to be a recipe book for clinical molecular tests, simply because the specifics of testing change quite rapidly in molecular pathology as new technologies emerge and are integrated into clinical molecular practice. Instead, the emphasis remains the molecular variants being detected for clinical purposes, the clinical usefulness of molecular test results, and the clinical and laboratory issues that require special attention. While this textbook focuses on molecular and genomic testing, with only a single chapter covering proteomics, the reader must understand that the genome does not drive all disease and health, but works in concert with the environment, the metabolome, the methylome, and other determinants of disease and health.

As we move toward genomic medicine, the molecular pathologist and all pathologists will play a significant role in the proper utilization of molecular and genomic tests to improve patient outcomes and the cost-effectiveness of the care we deliver. In the era of US healthcare reform, the promise of genomic medicine aligns almost perfectly with the healthcare reform goals of improving individual patient outcomes, improving the health of populations, and reducing the cost of healthcare. While much of genomic research focuses on the clinical

significance of pathogen and patient genomic variants for diagnosis and therapy, evidence of the value of genomics in clinical care also is needed, especially as we move toward population health management and global payment models.

My hope is that you apply the information in *Molecular Pathology in Clinical Practice* to the care you provide for your patients.

Burlington, VT, USA Debra G.B. Leonard

http://www.springer.com/978-3-319-19673-2

Molecular Pathology in Clinical Practice
Leonard, D.G.B. (Ed.)
2016, XIX, 1001 p. 219 illus., 132 illus. in color.,
Hardcover
ISBN: 978-3-319-19673-2