Preface

The specialty of molecular genetic pathology (MGP) is rapidly growing and evolving. It focuses on the molecular identification of inherited genetic conditions, of acquired genetic diseases such as solid tumors and hematologic malignancies, and of infectious diseases. Specialty board examinations in MGP are available to physicians who are pathologists or medical geneticists and who have completed subspecialty training in an accredited MGP training program. Prior to the conception of MGP, specialty board examinations were already administered in clinical molecular genetics (CMG), which requires training by M.D. or Ph.D. post-doctoral trainees. CMG training programs focus specifically on inherited genetic conditions. The intended audience for this text comprises trainees in MGP and CMG, as well as residents and fellows in medical specialties to which molecular genetic pathology is pertinent. It is also relevant to the practicing pathologist who wants to learn more about the current practice of molecular diagnostics.

In the past few years, much needed reference textbooks have become available and provide a terrific knowledge foundation and resource. The book in your hands takes a complementary approach. It is a practical, completely case-based book with examples of molecular diagnostic cases (which are composites with fictitious patient names), as they can be encountered in molecular pathology laboratories. The cases are divided into the four main areas addressed in MGP: inherited conditions, hematopathology, solid tumors, and infectious diseases. Each section includes topics ranging from test selection, qualitative and quantitative laboratory techniques, test interpretation, and prognostic and therapeutic considerations, to ethical considerations, technical troubleshooting, and result reporting. This reflects a rich variety of teaching points associated with the diversity of cases in molecular laboratories and represents a cross section of current practical issues which are encountered in the day-to-day activities of a molecular genetic pathologist. The scenarios presented are not intended to indicate the preferred or only approach, but rather represent examples of current practice. Some of the cases in this book reflect common scenarios, whereas others are complex "puzzlers." All provide an opportunity to actively engage with the presented material and to independently develop approaches, solutions, and diagnostic interpretations. As such, it is a practice-based preparation for board examination, for the extensive range of clinical scenarios in the medical specialty of MGP, and, most importantly, for its successful practice.

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