Preface

The recent development of high-throughput next-generation sequencing (NGS) technology has transformed the way DNA-based molecular diagnostic testing is performed in clinical laboratories. NGS allows parallel sequencing analyses of multiple genes effectively at any desirable depth of coverage. It is often difficult for clinicians to fully understand and utilize NGS-based tests effectively due to the sophisticated instrumentation, the enormous amount of data generated, the complex analytical tools involving sequence alignment, and the bioinformatics for variant annotation. While professional interpretation can address these issues, the purpose of this book is to help physicians to better understand the science behind and clinical utility of NGS to maximize benefit for their patients.

In Part I, DNA sequencing principles, the underlying chemistries, and the development of NGS technology are described. This is followed by an overview of the methods used in the traditional molecular diagnosis of human genetic diseases. Part II details the instrumentations, bioinformatics, and computational techniques involved in NGS. This section of the book includes a comparison of the different methods used for target gene enrichment and the various sequencing platforms, as well as the algorithms and bioinformatics used for sequence analysis, variant annotation, and interpretation of the final results. Part III describes specific applications of NGS to the molecular diagnosis of clinically defined diseases (e.g., congenital disorders of glycosylation), genetically heterogeneous disorders involving many different genes leading to the same or similar clinical phenotypes (e.g., retinitis pigmentosa), diseases associated with a particular region or a whole chromosome (e.g., X-linked intellectual disability), and dual genome mitochondrial respiratory chain disorders involving nuclear and mitochondrial DNA. The utility of NGS for noninvasive prenatal diagnosis is also described. Part IV discusses the College of American Pathologists (CAP) / Clinical Laboratory Improvement Amendments (CLIA) guidelines for establishing a clinically based test using novel technology and compliance issues for laboratories offering NGS-based testing.

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