

Preface

Recent development of high-throughput next generation sequencing (NGS) technology has transformed the way DNA-based molecular diagnostic testing is performed in clinical laboratories. In the past few years, clinically validated NGS has been applied to routine molecular diagnosis of human genetic diseases. In this book, we review the outcome of NGS in clinical practice.

Pitfalls of traditional PCR-based Sanger sequencing can be overcome by NGS. This book first reviews the technologies of NGS and their advantages over traditional Sanger sequencing and why NGS has become the new gold standard for clinical molecular diagnosis. In addition, clinically validated deep NGS can accurately detect not only single nucleotide variants (SNVs) but also copy number variants (CNVs). The ability to simultaneously detect and quantify SNVs and CNVs in multiple genes makes NGS an ideal comprehensive approach for molecular diagnosis. Applications of NGS to genetic analysis of various disease areas, such as metabolic disorders, retinal disease, hearing impairment, primary immunodeficiency, bone disorders, hereditary cancer, RASopathies, complex neuromuscular disorders, diabetes, cardiovascular genetic diseases, mitochondrial DNA-related disorders, whole exome sequencing in clinical setting, family-based studies, and carrier screening, are described in individual chapters. The complete coverage of clinical utility of NGS is of course beyond the scope of this book. We hope that through the examples described in this book, readers will have an overview of how NGS is utilized in clinical diagnostic laboratories.

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