SPRINGER BRIEFS IN GENETICS

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Next Generation Sequencing Technologies in Medical Genetics

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Preface

The purpose of this book is to serve as an introduction to those that want to learn about next-generation–sequencing (NGS) and its applications. However, from the middle to end of each chapter a more in-depth approach will be taken to satisfy the curious minds of professionals in the fields of medical genetics and other related disciplines. Most chapters will contain one figure or table illustrating principles or summarizing key findings in the field.

A book search on Amazon and Google on "next-generation–sequencing" leads to book hits covering a wide range of topics including (1) NGS methods, (2) NGS informatics, (3) application to microRNA expression profiling, (4) application to personalize medicine, (5) application to plant sciences, and (6) challenges and opportunities of NGS for biomedical research. However, none of these books describes the direct application of NGS to medicine, specifically, laboratory medicine or molecular diagnostics. This book will bridge the gap between research and direct application to patient care. I foresee this book as the first, of many to come, translation medicine books in this field. Moreover, being a part of the Briefs in Genetic series will allow the reader to quickly become familiar with the technologies and most importantly their clinical applications. Furthermore, throughout the book the recent developments are briefly summarized.

In this book (Part I), we introduce the reader to the wealth of next-generation technologies followed by their direct applications (Part II) in the diagnosis of genetic disorders in the field broadly known as medical genetics. We will separate Parts I and II equally because this will allow a more in-depth description of the technologies for those that require a more profound understanding of the technologies. The equal space split, Part II, will provide the opportunity to describe the applications of NGS to molecular diagnostics by starting with a comprehensive view of the genetic disorders that have been analyzed by these technologies and then we focus on several of these genetic disorder examples including muscular dystrophy and hearing loss. Within these disorder-based chapters, we will describe the disorder and how NGS has been an excellent tool for reaching a diagnosis of previously undiagnosed patients. Furthermore, we will discuss the additional NGS

benefits, namely, increased sequencing throughput and decreased cost that patients can obtain from these tests. It can be speculated that NGS will become an even more popular platform in laboratory medicine and it can be argued that the technology is here to stay to provide better patient care by reaching a diagnosis sooner. Finally, we will end the book by briefly acknowledging the breakthroughs, in light of eight other chapters that describe the triumphs of the technologies, and focus on the challenges that lie ahead and suggest possible solutions to such challenges.

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This book is dedicated to our families, who supported and encouraged us in spite of all the time it took me away from them.

C. Alexander Valencia, Ph.D.