

Preface

The introduction of next-generation sequencing (NGS) technologies revolutionized the means by which scientists extract genetic information from biological systems, and revealed the virtually limitless insight that can be gained from the genome, transcriptome, and epigenome of various species. Following the lead of the groundbreaking Human Genome Project, several additional large-scale genome studies have materialized all over the world with NGS being the cornerstone of these investigations. Furthermore, the application of modern NGS techniques in the clinical field has led to major breakthroughs in the identification of disease-related genes in various forms of cancer and other life-threatening ailments.

A remarkable feature of NGS technologies is their high throughput nature which results in hundreds of thousands or even millions of short-read sequences. Therefore, research in the NGS field is interdisciplinary and requires integration between biological and computational knowledge and skills. In fact, the analysis of NGS data requires intensive computational power and skillful bioinformatics personnel.

However, NGS is not without its own challenges, requiring continuous development in sequencing technologies, computational infrastructure, and bioinformatics techniques to analyze the resultant raw data in order to assemble and annotate the full-length genome and transcriptome. Such developments have led to remarkable progress in efforts to enhance the performance and coverage of sequencing, and yielded a dramatic improvement in the quality of assembled sequences. Nevertheless, issues such as short-read lengths, sequencing and platform-specific errors, and large-scale memory requirements for the assembly process remain major challenges in the field.

As part of the SpringerBriefs series, this book presents a brief overview of the history, development, methods, applications, and challenges of NGS, and is divided into three parts. Part I provides an introduction to the basics of molecular biology, algorithms, and data structures required to assist readers in understanding the more technical portions of this book (Chaps. 1 and 2). Part II discusses NGS methods and the associated platforms, applications, challenges, and recent advancements

(Chaps. 3–7). Lastly, Part III provides an overview of NGS assembly stages and the related assessments and evaluations, utilized tools and remaining challenges in the field (Chaps. 8–11).

The primary audience intended for this book is newcomers to the field of sequencing with either a biological or computer science background. We provide basic introduction to both these scientific areas in relation to sequencing to allow readers to appreciate the unique amalgamation between the two that has pushed forward modern developments in the area. Furthermore, the book will also be useful for readers with a seasoned background in sequencing, as Parts II and III include a comprehensive topical review of the field including discussions on prevailing stumbling blocks in relation to technical complications, widespread availability, and the continuing need for various resources. In addition, Chap. 10 will present a unique glimpse into the recent, yet rapidly developing, field of the assessment of the next-generation sequence assembly.

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