

Preface

The completion of the human genome, with its more than 3 billion base pairs (bp) of sequenced DNA, has provided an unprecedented wealth of knowledge. With the additional investigation of single nucleotide polymorphisms (SNPs), we have also learned how little genetic variability there truly is in the human genome. Moreover, genome-wide association studies (GWAS) have revealed important genotype-phenotype correlations. Nevertheless, our understanding of the functionality of the genome is still lacking.

Dispersed among its 3 billion bp, the human genome features approximately 20–25,000 functional genes that encode various proteins and their isoforms. In recent years, however, scientists realized that the functionality of the genome is not restricted to only protein-encoding genes, which are transcribed into messenger RNAs, but also to the transcription of non-coding RNAs [e.g., microRNAs (miRNAs)], which play important roles in the posttranscriptional control of gene expression and, consequently, influence the resulting phenotypes.

Broadly speaking, it is at this point—from studies investigating where the functions of the genome first begin—that the science of transcriptomics emerged. For example, how are RNA molecules transcribed, what are the different species of RNA, what are the functions of each of these species and how are they differentially expressed among cells, tissues and organs?

Transcriptomics can therefore be thought of as the molecular biology of gene expression on a large scale. It is derived from functional genomics studies with a focus on transcription. Since its inception, transcriptomics has benefitted from and will continue to benefit from microarray technology. Sequencing is undoubtedly the ultimate tool when the objective is to delve into the differences at the sequence level or to confirm the specific RNA isoform involved. Even more so now, with the emergence of new technologies for high-throughput RNA sequencing (RNA-Seq), we can answer more questions about the structure of RNAs, such as those found in alternative splicing. However, the bottleneck remains in the data analysis because sequences are currently being obtained in quantities that have never been previously achieved.

However, as microarray bioinformatics has reached a very advanced stage (with more than 15 years to perfect the analysis pipeline) and as microarray slides them-

selves have become increasingly “large”, currently encompassing sequences from the entire functional genome plus the complete set of known non-coding RNAs, researchers have not neglected the applications of this important technology.

Recent comparative analyses have indicated a strong concordance between exon microarrays and RNA-Seq data. Therefore, the goal is now to use these two complementary strategies for in-depth transcriptomics studies.

This book was organized on the basis on these assumptions. It includes 17 chapters and covers the fundamental concepts of transcriptomics, as well as the current analytical methods. We provide examples in high-level technical and scientific detail, using accessible language whenever possible, as each chapter is written by experienced and productive researchers in the field.

Over the first six chapters (Part I), we introduce the concept of the transcriptome, as well as how microarrays or RNA-Seq can be used to trace expression signatures, measure transcriptional expression levels and establish connections between genes based on their transcriptional activity in normal cells, differentiating cells and organs.

Chapters 7–17 (Part II) then provide examples of the state of the transcriptome associated with major human diseases, such as inflammatory diseases, autoimmune diseases, metabolic diseases (such as type 2 diabetes mellitus), genetic diseases (such as Down syndrome), cancer and infections caused by pathogenic microorganisms, such as tuberculosis mycobacteria, fungi and the protozoan *Trypanosoma cruzi*, which is the causative agent of Chagas disease.

Special attention is also given to Chap. 17, which was strategically placed at the end of this book. The author of this chapter, who was one of the original developers of microarray technology in the mid-1990s, discusses the medical potential of transcriptomics from an analytical point of view.

I hope this book will be useful to researchers who wish to gain a comprehensive view of transcriptomics in health and human disease. I would like to thank all of the authors for their dedication and time spent writing these chapters. Finally I thank Springer for providing this opportunity and for its continued support during the writing and organization of this work.

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