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## Preface

The ultimate goal of the Human Genome Project is to understand the biology and underlying physiology of human health and disease. Functional genomics has become one of the major focuses in molecular biology in this post-genomics era. It is no longer difficult to initiate DNA variant screening at a genome scale by using massive parallel sequencing technologies. However, it remains a challenge to decode and understand hundreds, indeed thousands, of identified variants. Also, what constitutes a gene has been dramatically expanded by the discovery of widespread transcription beyond the protein coding unit. Therefore, we need more sophisticated tools to assist us in the identification of the functionality of undefined genes and the correlation of DNA variants with a particular phenotype.

In silico tools are pivotal along the journey of gene discovery. Although there is a wide spectrum of these tools, they are not well disseminated or easily applied by the end users. The scope of this volume of *Methods in Molecular Biology*<sup>TM</sup> is to collect common and useful in silico tools. These tools have been arranged into three sections. The first section (**Chapters 1, 2, 3, 4, 5, 6, and 7**) includes locus mapping information on linkage analysis, association mapping, integrative analysis, and exome analysis. Tools for DNA marker selection, in silico PCR, and statistical analysis are also provided in Section I. The second section (**Chapters 8, 9, 10, 11, and 12**) focuses on gene discovery from a defined locus. Included are in silico tools for knowledge tracking, application of gene ontology, phenotype mining, and in silico gene prioritization. Finally, in the last section (**Chapters 13, 14, 15, 16, 17, 18, 19, 20, and 21**), many useful in silico tools are presented for the functional characterization of genes, which includes DNA sequencing analysis, variant characterization, as well as RNA structure prediction. Detailed protocols are provided for the design and analysis of quantitative PCR, and the prediction of both transcriptional factor-binding sites and potential splice-affecting DNA variants. In silico tools are also assessed for the prediction of post-translational modifications as well as protein motif discovery and structure analysis.

Each chapter provides a brief introduction and clear instructions for the applications of a particular in silico tool. Furthermore, each chapter is supplemented with additional notes that provide insights into the working of the tool in question. These brief notes provide valuable keys that allow for successful application. Hopefully researchers in the field of gene discovery will find this series of articles resourceful and easy to follow. The applications of these in silico tools will facilitate locus mapping, accelerate gene identification, and help ascertain the functionality of DNA variation.

We would like to express our gratitude to all contributors of this book *In Silico Tools for Gene Discovery* for their collaboration and collective effort. We also thank our series editor, Professor John Walker, for his help and guidance throughout the process.

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