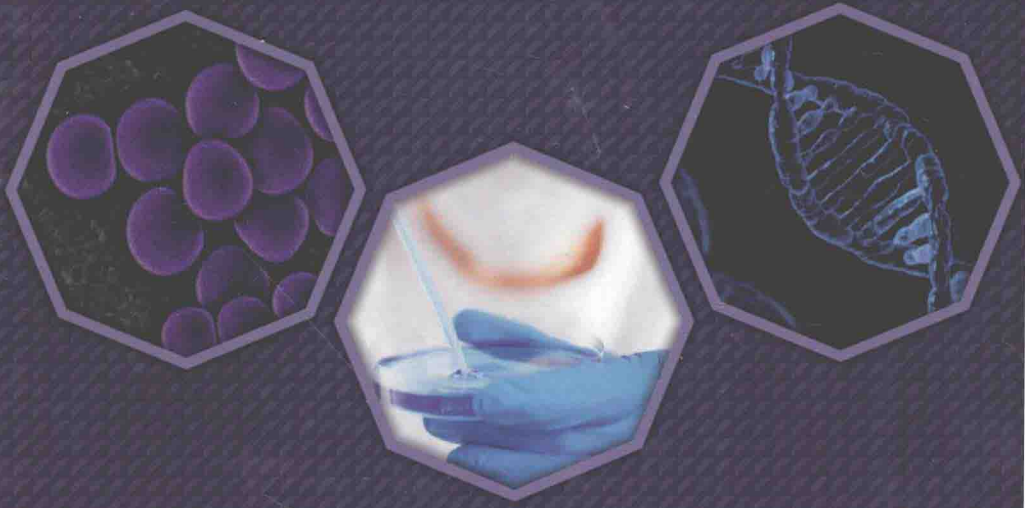
# OMICS in Clinical Practice

## Genomics, Pharmacogenomics, Proteomics, and Transcriptomics in Clinical Research



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*Edited by*

**Yu Liu, PhD**



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# OMICS IN CLINICAL PRACTICE

## Genomics, Pharmacogenomics, Proteomics, and Transcriptomics in Clinical Research

# ABOUT THE EDITOR

**YU LIU, PhD**

As a bioinformatician, Dr. Yu Liu's research is centered on the development and application of computational tools for the study of complex diseases. He has extensive experience with data generated from microarrays, next generation sequencing and high-resolution mass spectrometry, and developing bioinformatics tools and applying system biology approach to study complex diseases, like sleep apnea, neurodegenerative diseases, and cancers. More recently, he developed a systems biology approach that enables the discovery of high-level disease mechanisms and provides testing hypotheses for further research. Currently, Dr. Liu is a senior research associate at the Center for Proteomics and Bioinformatics at Case Western Reserve University, Cleveland, Ohio. He received a PhD in Bioinformatics from Montreal University, Montreal, Canada, and has postdoc training from the University of Toronto, Canada.

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The editor and publisher thank each of the authors who contributed to this book, whether by granting their permission individually or by releasing their research as open source articles or under a license that permits free use provided that attribution is made. The chapters in this book were previously published in various places in various formats. To cite the work contained in this book and to view the individual permissions, please refer to the "How to Cite" notes at the beginning of each chapter. Each chapter was read individually and carefully selected by the editor. The result is a book provides a comprehensive introduction to genomics, proteomics, and transcriptomics in relation to human health and disease.

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# INTRODUCTION

This book serves as an introduction to genomics, proteomics, and transcriptomics, putting these fields in relation to human disease and ailments. The various chapters consider the role of translation and personalized medicine, as well as pathogen detection, evolution, and infection, in relation to genomics, proteomics, and transcriptomics. The topic of companion diagnostics is also covered.

The book is broken into five sections. Part I examines the connection between Omics and Human disease. Part II looks at the applications for the fields of translational and personalized medicine. Part II focuses on molecular and genetic markers. Part IV describes the use of omics while studying pathogens, and Part V examines the applications for companion diagnostics.

Mitochondria are the most complex and the most important organelles of eukaryotic cells, which are involved in many cellular processes, including energy metabolism, apoptosis, and aging. And mitochondria have been identified as the "hot spot" by researchers for exploring relevant associated dysfunctions in many fields. In Chapter 1, the emergence of comparative proteomics enables Jiang and Wang to have a close look at the mitochondrial proteome in a comprehensive and effective manner under various conditions and cellular circumstances. Two-dimensional electrophoresis combined with mass spectrometry is still the most popular techniques to study comparative mitochondrial proteomics. Furthermore, many new techniques, such as ICAT, MudPIT, and SILAC, equip researchers with more flexibilities inselecting proper methods. This article also reviews the recent development of comparative mitochondrial proteomics on diverse human diseases. And the results of mitochondrial proteomics enhance a better understanding of the pathogenesis associated with mitochondria and provide promising therapeutic targets.

Omics approaches to the study of complex biological systems with potential applications to molecular medicine are attracting great interest in

clinical as well as in basic biological research. Genomics, transcriptomics and proteomics are characterized by the lack of an a priori definition of scope, and this gives sufficient leeway for investigators (a) to discern all at once a globally altered pattern of gene/protein expression and (b) to examine the complex interactions that regulate entire biological processes. Two popular platforms in "omics" are DNA microarrays, which measure messenger RNA transcript levels, and proteomic analyses, which identify and quantify proteins. Because of their intrinsic strengths and weaknesses, no single approach can fully unravel the complexities of fundamental biological events. However, an appropriate combination of different tools could lead to integrative analyses that would furnish new insights not accessible through one-dimensional datasets. In Chapter 2, Silvestri and colleagues outline some of the challenges associated with integrative analyses relating to the changes in metabolic pathways that occur in complex pathophysiological conditions (viz. ageing and altered thyroid state) in relevant metabolically active tissues. In addition, the authors discuss several new applications of proteomic analysis to the investigation of mitochondrial activity.

The wide application of next-generation sequencing (NGS), mainly through whole genome, exome and transcriptome sequencing, provides a high-resolution and global view of the cancer genome. Coupled with powerful bioinformatics tools, NGS promises to revolutionize cancer research, diagnosis and therapy. In Chapter 3, Shyr and Liu review the recent advances in NGS-based cancer genomic research as well as clinical application, summarize the current integrative oncogenomic projects, resources and computational algorithms, and discuss the challenge and future directions in the research and clinical application of cancer genomic sequencing.

The mapping of the human genome and subsequent advancements in genetic technology had provided clinicians and scientists an understanding of the genetic basis of altered drug pharmacokinetics and pharmacodynamics, as well as some examples of applying genomic data in clinical practice. This has raised the public expectation that predicting patients' responses to drug therapy is now possible in every therapeutic area, and personalized drug therapy would come sooner than later. However, debate continues among most stakeholders involved in drug development