METHODS IN MOLECULAR BIOLOGY

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Precision Medicine

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Preface

With the development of omics technologies, especially next-generation sequencing, disease progression can be investigated in an unprecedented way. The multi-omics approaches reveal the essence of disease pathology and make precision diagnosis and therapy possible. Precision medicine will transform the medical practice fundamentally. In these 19 chapters, we will introduce the technologies and applications of precision medicine.

The first four chapters present emerging experimental and bioinformatics technologies that are widely used in precision medicine, such as T cell receptor repertoire sequencing, nanopore sequencing, microsatellite instability, and network analysis. The T cell receptor repertoire sequencing can help monitoring immune responses and predicting the prognosis of disease. Nanopore sequencing as third-generation sequencing has great potential for point-of-care testing (POCT) due to its fast turn-around time, portable and real-time data analysis. Microsatellite instability is a key indicator for predicting response to anti-PD-1 inhibitors. Network analysis can integrate multi-omics big data and reveal the molecular mechanisms underlying complex diseases.

The next six chapters are focused on cancer studies. There have many successful applications of precision medicine in cancers. Breast cancer, prostate cancer, lung cancer, nasopharyngeal carcinoma, and cancer of unknown primary origin are discussed. Biomedical imaging, methylation, immunohistochemistry, and gene expression can all be used for cancer diagnosis and treatment optimization. Even cancer of unknown primary origin can be traced based on their molecular characteristics.

Cardiovascular diseases (CVDs) are the most common diseases and cause many deaths each year widely. There are five chapters for cardiovascular diseases, such as atrial fibrillation (AF), atherosclerosis, dilated cardiomyopathy, and total anomalous pulmonary venous connection (TAPVC). DNA methylation has potential value in being biomarkers and underlying the diagnosis and prognosis of atrial fibrillation. Proprotein convertase subtilisin/kexin type 9 (PCSK9) plays an important role in atherosclerosis and shows therapeutic potentials.

The next two chapters are focused on other complex diseases beside cancers and cardiovascular diseases, such as chronic obstructive pulmonary disease (COPD) and systemic lupus erythematosus (SLE). Chronic obstructive pulmonary disease (COPD) is a common disease with high morbidity and mortality in the world. Airway inflammation biomarkers can facilitate precise manage of neutrophil-predominant COPD. Systemic lupus erythematosus (SLE) is a complex autoimmune disease which faces difficulties in treatment. Stratification of SLE patients based on genetic profiling will enable us to make more effective and precise choices for treatment plans.

The last two chapters introduce the latest engineering and surgical developments in precision medicine. Scientists seek various engineering approaches, such as 3D printing, to harness stem cells, scaffolds, growth factors, and the extracellular matrix to promise enhanced and more reliable bone formation. Ex Vivo Lung Perfusion (EVLP) is a technique for extending lung preservation time and repairing lung injury in the field of lung transplantation. EVLP can increase the number of lungs that meet the transplant criteria and, to some extent, alleviate the current shortage of donor lungs.

As mentioned above, this book covers most perspectives of precision medicine, from basic research to clinical surgeries, from cancer to cardiovascular disease, from sequencing technology to big data analysis. It is hoped that this book can broaden the horizons for researchers, engineers, and clinicians and accelerate the interdisciplinary precision medicine research and applications.

Shanghai, China *Tao Huang*

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