Preface

Epigenetics is an emerging frontier of biology, and its definition is continuously being adapted based on new scientific findings. In fact, the NIH Roadmap Epigenomics Project has recently defined epigenetics as "the heritable changes in gene activity and expression (in the progeny of cells or of individuals) and also stable, long-term alterations in the transcriptional potential of a cell that are not necessarily heritable." In this regard, epigenetics includes DNA methylation, noncoding RNAs, and histone posttranslational modifications. This integrative definition of epigenetics reflects the potential of the discipline to expand beyond the control of a particular gene expression program for each cell type, defining the cellular and developmental identity and function of cells, and, finally, translating this potential to health and disease conditions in human beings.

Due to the rapid progress in the field of epigenetics, new and promising methodologies to advance biomedical research are being developed. Epigenetic research and epigenetic pharmaceutical drug development are now considered areas of great interest and promise in the biomedical scene. The advantage of human epigenetics compared with human genetics and genomics is that it provides vital information about gene function in individual cell types, while incorporating information from the environment and lifestyle, and unlike most genetic defects causative of human disease, epigenetic alterations are modulable and reversible. The volume *Epigenetic Biomarkers and Diagnostics* is intended to describe both epigenetic biomarkers that can be adopted into clinical routine as well as advanced technologies and tools for their analysis. In this regard, epigenetic biomarkers provide clinicians valuable information about the presence or absence of a disease (diagnostic value), the patient prognosis (prognostic value), the response to a specific treatment (predictive value), the effects of ongoing treatment (therapy-monitoring biomarkers), and the future risk of disease development (risk prediction). Furthermore, several advantages may arise from the use of epigenetic biomarkers versus gene expression in clinical practice, such as higher stability, for example, in biofluids. They can also fill clinical gaps by bridging genetic information, mRNA transcription, and protein translation. In consequence, the associations between epigenome alterations and diseases become clearer, providing a way to act directly on gene expression by developing specific drugs or even by adopting healthy lifestyles.

Many methodologies, including classical methods and next-generation-sequencing-based technologies, are available to clinicians and researchers to identify new epigenetic biomarkers and analyze them from several biological sources. In this context, genome-wide methylation analysis, chromatin immunoprecipitation coupled with high-throughput platforms, and noncoding RNA sequencing are described in this volume. Furthermore, some techniques for DNA methylation analysis are more likely to be rapidly adopted in clinical laboratories, such as EpiTYPER MassARRAY, methyl specific PCR (MSP), and pyrosequencing. On the other hand, immunoassays are well established in

clinical laboratories for the analysis of histones (i.e., inflammatory and autoimmune diseases). However, it is expected that the incorporation of mass spectrometry technologies into laboratories for clinical diagnostics (replacing routine immunoassays) will be the tendency in the coming years, as will be the analysis of histone posttranslational modifications associated with pathological states.

Although it is not possible to cover all epigenetic markers, this volume includes chapters describing the most promising biomarkers for cancer (i.e., breast, lung, colon, etc.), metabolic disorders (i.e., diabetes and obesity), autoimmune diseases, infertility, allergy, infectious diseases, and neurological disorders; and, where possible, we will focus our attention on those which are feasible to be adopted for clinical use.

This book was written in a comprehensive manner by outstanding experts in their corresponding fields for a broad target audience such as advanced students, basic scientists, biomedical and biotechnological companies, as well as clinical researchers, clinicians (i.e., pathologists, immunologists, oncologists, endocrinologists, etc.) and analysts from clinical laboratories who can adopt these potential biomarkers into clinical practice.

In the coming years, epigenetics will continue to provide an exciting future in biomedicine and clinical practice. The chapters covered in *Epigenetic Biomarkers and Diagnostics* highlight the unprecedented impact of epigenetics in clinical diagnostics and will contribute to the discovery and development of new epigenetic biomarkers in the future.

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