Foreword

Molecular epidemiology provides an exciting set of opportunities to contribute to the evidence base for the prevention of chronic diseases in the coming decades. In the mid- to late-1980s, the emergence of the polymerase chain reaction resulted in a step-change in the ability to investigate genetic polymorphisms and disease risk. This area was further transformed by the Human Genome Project and the widespread application of genome-wide association studies to large, multicentre case–control studies. Nevertheless, I believe the best is still to come from molecular epidemiology. This assertion is based on a combination of advances in understanding molecular mechanisms underlying disease development, powerful new laboratory technologies to interrogate patterns of gene, protein and metabolite levels, and their potential application to biobank specimens associated with large-scale population-based cohort studies.

There are risks to the fulfilment of this promise. First, the exquisite tools to study genetic susceptibility are as yet unmatched by tools of equal power to evaluate the environmental (non-genetic) basis of disease; without a balance between the genome and the exposome, their interplay in the causation of chronic diseases cannot be fully elucidated. Second, a systematic investment by research organizations and funders is needed in the type of translational research that draws advances in mechanistic knowledge and the associated technologies into epidemiology; interdisciplinary research across the laboratory sciences, epidemiology, clinical research, biostatistics and bioinformatics has never been more important.

This IARC publication, prepared by experts in the field, is a timely and valuable foundation for the future. It emphasizes the development and validation of appropriate methodology. It highlights the flow of knowledge from mechanisms of disease development, through the derivation of biomarkers, to their application in epidemiological studies. It illustrates the benefits of mentally crossing disease boundaries when considering the origins and consequences of underlying pathological processes. It stimulates inter-disciplinary thinking and orientates the laboratory towards public health.

The book spans great scale, highlighting at one end of the spectrum the increasing requirement to handle and interpret through computational means tens of millions of biomarker data points on tens of thousands of subjects while, at the other end, being attentive to the ethical questions affecting the individuals contributing to research through donation of their time, information and biological samples. If molecular epidemiology is to truly contribute to relieving the ever-increasing burden of chronic disease it will need excellent communication not only to the scientific audience that is the target of this book, but the people worldwide who are the subject of its investigations and concerns.

Christopher P. Wild
Director, International Agency
for Research on Cancer

Major advances in our understanding of the origins and natural history of several chronic diseases have come from epidemiologic and laboratory research over the past 1–2 decades. While this knowledge has provided new opportunities for disease prevention and control, we are still limited by an incomplete grasp of causal mechanisms, which hold the key to further progress in preventive medicine and public health. However, recent conceptual breakthroughs in genomic and molecular sciences have fuelled optimism that the incorporation of innovative high-throughput technologies into robust epidemiologic designs will further dissect the genetic and environmental components underpinning complex diseases such as cancer, and thereby inform new clinical and public health interventions.

At this critical moment in the evolution of molecular epidemiology, the editors of this volume have enlisted scientific leaders in the field to review the major concepts, methods and tools of this interdisciplinary approach. The chapters summarize recent progress that has been made for several diseases and traits through molecular epidemiology, while suggesting promising directions for further discovery. Special attention is given to the process of selecting, validating and integrating molecular and biochemical biomarkers that sharpen our measures of causal factors and mechanisms, as well as disease outcomes, through epidemiologic research. The success of molecular epidemiology is due in no small part to advances in statistical methods and bioinformatics, as illustrated by the discovery of heritable mechanisms for many diseases and traits generated recently by large-scale genome-wide association studies.

As a fast-breaking interdisciplinary approach, molecular epidemiology faces formidable challenges, but the dividends are likely to increase by an order of magnitude as the next-generation "omics" technologies become available for epidemiologic application. With the evidence in this volume as a starting point, the stage is set for basic, clinical and population scientists to accelerate collaborative efforts that will contribute new biological insights and augment strategies for preventing and controlling disease on a global scale.

Joseph F. Fraumeni, Jr.
Director, Division of Cancer Epidemiology and Genetics, National Cancer Institute