Preface

The scope of the field of genomics has expanded rapidly with the completion of the human genome sequencing project. Current understanding of biological systems has changed dramatically due to the combination of new technologies and the amount of data available, allowing for experimentation on a scale previously unimaginable.

The post-genomic era has opened up a plethora of opportunities for academic and commercial exploitation of these novel technologies. As increasing numbers of investigators seek to harness the fruits of genomics knowledge, it is essential that well-tested protocols are made available to researchers. With this in mind, it is apposite to launch this collection of protocols written by experts who are routinely employing these techniques in their laboratories.

This book represents more than a collation of step-by-step laboratory techniques, as it outlines the concepts underlying the techniques, and where possible provides alternative methods. This aspect adds considerable value to this book, and differentiates it from other 'protocol books'. All contributing authors have taken great care in explaining the principles of the techniques described, prior to presenting a detailed protocol.

Understandably, this volume does not purport to be a comprehensive collection of protocols in genomics per se; instead, the focus has been placed on key techniques in genomics and its derivative disciplines. The range of techniques presented is broad, and includes the detection of genetic variation, mRNA and genomic DNA copy number profiling, analysis of proteins by experimental and *in silico* methods, and the application of genomic strategies for therapeutic intervention.

Chapters 1–3 present procedures for genome analysis, including alternative strategies for the detection of chromosomal copy number alterations. The anomaly that it is often difficult to collect fresh tissues for research, and yet huge tissue banks exist in histopathology departments, is acknowledged by the description of procedures able to use archival samples (Chapters 1 and 3). The identification of single nucleotide polymorphisms (Chapter 2) has been instrumental to strategies for high-resolution, genome-wide association analysis (Chapter 4) in studies of disease susceptibility and pharmacogenetics.

Techniques for the analysis of gene expression are presented in Chapters 5–7. The trend in transcriptome analysis is towards the profiling of more strictly defined cell populations, necessitating the use of RNA amplification techniques that are discussed in Chapter 5. Real-time quantitative reverse transcription–PCR techniques (Chapter 6) are widely used for the detection and quantification of RNA as a consequence of their sensitivity and specificity. At the other end of the spectrum, there are many applications that require the capability to express transgenes *in vivo*, and Chapter 7 describes approaches for facilitating this via novel gene transfer methods.

The study of protein–protein interactions assists the understanding of biological function and elucidation of biochemical pathways, and Chapter 8 details use of the yeast two-hybrid system to generate high-confidence binary protein interaction data. The determination of gene function is central to functional genomics, and Chapters 9 and 10 explain alternative strategies towards attaining this objective.

The use of gene therapy for the modification of defective genes associated with pathogenesis is increasingly considered to be a viable approach for the treatment of disease. The penultimate two chapters address the issues involved and the strategies deployed.

Proteomic analysis is often a natural adjunct to transcriptional investigations, and the final chapter affords an introduction to protein profiling for the genomics specialist. This chapter is presented in a different format to the preceding chapters in that it offers a strategic guide, including a description of how to deal with some of the major issues and problems arising with protein profiling technologies.

We sincerely hope that these protocols, together with the summaries of their rationale, will help both experienced and new entrants in this field to carry out their experiments successfully.

Finally, we would like to thank all the contributing authors, David Hames, Clare Boomer and Jonathan Ray, the staff at Wiley-Blackwell, and most importantly our families for their support and endurance during the editing period.

Mike Starkey
Ramnath Elaswarapu