Preface

Since the release of the working draft of the human genome, there has been a need for improved technology and assay throughput to make use of the information now at our fingertips. *Pyrosequencing® Protocols* brings together world experts in fields where Pyrosequencing® has been an essential technology for achieving their goals.

The advantages of the Pyosequencing® system lie with the range of technological applications that can be applied to single nucleotide polymorphisms, insertion/deletions (indels), short tandem repeats, pooled allele frequencies, human leukocyte antigen typing, gene copy number, allelic imbalance in RNA, methylation status, and short sequencing stretches, many of which are described in this book. The assay is applicable to almost any source of DNA or RNA (e.g., blood, saliva, cell line, plasma, serum, tissue, formalin-fixed and/or paraffin-embedded samples, and whole genome-amplified DNA). In addition, the use of a universal biotinylated primer and multiplex analysis of up to three different amplicons can be performed, reducing genotyping cost and time of throughput. No other system provides this range, throughput, and cost advantage.

Within *Pyrosequencing® Protocols* the methods for utilizing Pyrosequencing technology are described in detail, including troubleshooting tips and background information. Chapter 1 provides an introduction to the fascinating origins of the methodology. Chapter 2 provides a brief overview of some of the applications Pyrosequencing® is used for. Chapters 3 and 4 describe primer selection and the basic technique. Chapters 5–7 provide methods for improving the throughput and decreasing the cost of Pyrosequencing®. Detailed applications for the technique can be found in Chapters 8–13, while the important aspect of data storage is discussed in Chapter 14.

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