Preface

Within the last decade, microarray technology has evolved from an emerging technology developed and used by a few laboratories into a well-established technology used in laboratories all over the world. In fact, the need to characterize genetic alterations is one of the highest priorities for the future of medicine and the clinical management of disease. This technology allows the rapid detection of point mutations, insertions or deletions, loss of heterozygosity, and gene amplification, which constitute the major nucleic acid variations associated with human disease. Additional disease-causing changes may involve DNA methylation and microsatellite instability for which automatable methods to detect instability in as few as 100 cells at multiple loci are required. Furthermore, in some instances it may be necessary to detect one tumor cell among a large number of normal cells, as well as to profile differentially expressed genes. Eventually, the ultimate goals are to characterize the entire genome rapidly and inexpensively, ideally using a single cell in order to survey the whole genome for any nucleic acid variation.

Within the field of proteome research, microarray technology has been adapted to the protein arena. Although DNA microarrays are quite popular and in vogue, proteins (not genes) are the targets for drugs; therefore, there is an increasing need to develop protein chips. Specifically, tools and methods are needed for the identification and quantification of proteins, and for the study of protein–protein interactions, enzyme–substrate interactions, and small-molecule interactions. Enormous efforts have been undertaken to transfer standard sandwich immunoassays in miniaturized and parallel formats to analyze simultaneously the expression of a large number of proteins, e.g., serum or tumor biomarkers.

It is becoming clear that microarray technology is capable of fulfilling these needs. Although some technologies are still confined to research laboratories, such as those aimed at performing resequencing of known genes and protein identification, rapid and robust methods are becoming available to address each of these needs. However, some general goals for diagnostics including sensitivity, specificity, high throughput, cost effectiveness, and turnaround time still need improvement.

Finally, it is also clear that new tools in the nanoscale format are on the horizon: quantum dots, nanoparticles, carbon nanotubes, and atomic force microscopes are now being used to directly probe DNA structure. These
technologies represent an emerging approach promising increased throughput, sensitivity, and sample processing, as well as facilitating single-cell and single-molecule detection.

Microarrays in Clinical Diagnostics offers an overview of the world of microarray technology. Because it is not clear which technology will eventually prevail, we have tried to assemble a comprehensive survey of the varied technologies now in use and to provide detailed methods sections in order to support scientists who design and perform microarray experiments.

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