Microarrays in Diagnostics and Biomarker Development

Current and Future Applications

Bearbeitet von
Bertrand Jordan

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Preface

This is the second time I assemble a book on microarrays for Springer—almost exactly 10 years after the first one (*DNA microarrays: Gene expression applications*. Springer Berlin Heidelberg 2001). During this interval, we have seen tremendous changes in the landscape of genomics and molecular biology. The first human genome sequence, with its revelations on gene number and relatively low human genetic diversity, was shortly followed by extensive investigation of single nucleotide polymorphisms in our DNA and, with the help of more and more complex "SNP arrays", by extensive whole-genome association studies (GWAS). These have uncovered many gene/phenotype correlations, but have also revealed how limited our understanding of the functional genome still is. The importance of non-coding transcripts and of epigenetic modifications of our DNA has been realised. And very recently, DNA sequencing technology has made tremendous progress, to the point that the "1,000-dollar genome" is in sight and that clinical use of whole-genome or exome sequencing is becoming significant (and is in fact competing with some uses of arrays). Meanwhile, clinical applications of DNA microarrays have developed, but not exactly in the directions anticipated 10 years ago and not quite at the level predicted by some analysts. Other microarrays using proteins, antibodies, peptide, or aptamer molecules as probes have made significant advances in spite of technical problems, and now have an impact not only in research but also in the clinic and in biomarker development.

This new book includes 13 main chapters, and covers essentially all types of arrays. It is focussed on entities that are in actual clinical use, or quite close to it—thus it does not discuss, for example, complex SNP arrays that remain essentially a research tool. It does present some very recent developments, such as peptide or aptamer arrays, or also miniaturisation towards "nanoarrays", that I see as having great potential in medicine even though their current presence is still limited. It discusses in detail very important issues in bioinformatics and in statistical analysis of array data, as well as the hurdles faced in the commercialisation of array-based tests and the vexing IP issues associated with these activities.

I believe this book will be useful to current array users who wish to have a complete view of the field, to newcomers who attempt to make the best choice between different technologies, and to academic scientists who engage in technology transfer activities and need to evaluate the hurdles involved in this process. I am especially grateful to the authors, my colleagues, who kindly accepted to undertake the absorbing task of writing a chapter, especially at a time where tight funding means that they are extremely busy preparing grant applications. And, of course, the organisational support from Springer to overcome the 1,001 small problems encountered in the production of this book is gratefully acknowledged.

Marseille, France Bertrand Jordan