
Cancer Diagnostics: Discovery and Clinical Applications—Introduction

Tumor marker analysis constitutes a sizable portion of routine clinical chemistry testing. This area of investigation has advanced considerably over the last 50 years. Many new tumor markers have been discovered and are now used routinely for cancer screening, diagnosis, and monitoring and for prediction of therapeutic response. Despite these advances, it is clear that the contribution of these markers to patient care and, especially, to altering clinical outcomes is relatively limited. Most, if not all, of the markers that we use today are compromised by their low diagnostic sensitivity and specificity. The nature of cancer as a disease is such that it is not acceptable to misdiagnose or mistreat patients. For this reason, tumor markers are not used for definitive diagnosis; they are used as aids to help physicians make decisions, after combining other clinical and diagnostic data.

Recent advances in the field of biological science have sparked new interest in the area of cancer biomarkers. The sequencing of the human genome has provided (or will provide) fundamental structural information about all human genes. Having all the genes on the table allows us to systematically study them globally as candidate biomarkers for cancer or other diseases. In addition, the advent of high-throughput technologies, including cDNA microarrays and biological mass spectrometry, has allowed thousands of measurements to be performed in short periods of time. The development of powerful bioinformatic approaches, to combine this information into meaningful output, has further contributed enormously to these new technologies. It is thus natural for people who work in the field to feel optimistic that these new resources and technologies will likely facilitate the discovery of new cancer biomarkers with improved sensitivity and specificity.

It is now very clear that we are at a crossroads in this field of investigation. Academic institutions and pharmaceutical and diagnostic companies are using these high-throughput strategies to discover the cancer biomarkers of the future. During this period, we have seen an

interesting phenomenon. Pharmaceutical companies are now very keen to develop diagnostics because they want to use them to optimize and clinically validate their drug targets.

The purpose of all these efforts is not solely the discovery of new diagnostics. Much work is focusing on new classification schemes for cancer, based on molecular changes or alterations of gene expression. These data, it is hoped, will aid in devising new therapeutic strategies and individualized treatments that may be effective in subgroups, rather than in the whole patient population.

Over the last 2–3 years we have witnessed the publication of seminal papers that preliminarily show the power of these new techniques. This does not necessarily mean that any new cancer diagnostics have already been discovered. In fact, a careful review of the literature suggests that we have not as yet seen breakthroughs in either cancer diagnosis or classification. We have seen proofs of principle of new concepts, but the data need reproduction and careful clinical validation. Many researchers believe that the best cancer markers have already been discovered. It also appears that the most promising approaches for the future will be to use panels of cancer biomarkers, which can be combined with artificial intelligence algorithms to produce diagnostic, predictive, and classification information that is more powerful than ever before. It is very likely that these predictions will come true over the next 3–5 years.

Clinical Chemistry has been in the forefront of publishing new knowledge in the field of cancer diagnostics for many years. Seminal papers describing novel biomarker discovery, clinical evaluations, and development of new technologies for measuring such biomarkers have appeared frequently in the pages of this journal, and do so today. By publishing this “Special Issue”, the journal underlines its commitment to play a major role in publishing novel information on discovery, clinical evaluation, and technologic developments in this area of investigation. In designing the content of this issue, we decided

to include several minireviews as well as original research papers. A glance at the Table of Contents reveals that the minireviews, as well as the papers, are highly diverse. We hope that this issue will bring focus to this discipline and

will allow our readers to obtain a global perspective of the cancer diagnostics field. We are interested in publishing future special issues in other fields. The input of readers in this undertaking is welcomed and highly appreciated.

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