

Executive Summary

Molecular diagnostics has become a large segment of the overall in vitro diagnostics (IVD) industry, with approximately \$3 billion or more in clinical sales worldwide in 2008 for kits and reagents sold by diagnostic companies. In addition, many of the molecular diagnostic tests that are available today are not sold as kits or reagents, but rather are laboratory-developed tests that are offered as a testing service by clinical laboratories and/or by diagnostic companies that have their own Clinical Laboratory Improvement Amendments (CLIA)-certified laboratory for performing their own proprietary tests.

The first molecular diagnostic tests to reach the market were infectious disease tests, and infectious disease testing remains the largest segment of the molecular diagnostics market. However, other applications of molecular diagnostics in the fields of oncology, personalized medicine, and inherited disorders, and for predicting risk of disease, have emerged, and these market segments are now growing rapidly. This report focuses on these other rapidly growing and emerging new applications of molecular diagnostics, as this field expands beyond infectious disease testing.

Chapter 2 briefly discusses selected technologies that are being used by molecular diagnostic companies. These include the early nonamplified probe-based assays, cytogenetic assays including fluorescent *in situ* hybridization (FISH), target and signal amplification technologies, multiplex technologies (including microarrays and others), and selected other current and emerging technologies.

Chapter 3 discusses the wide range of molecular diagnostic tests that are currently being performed, and also emerging new tests, in the field of oncology and other noninfectious disease fields. Oncology tests can be used for many different indications, including screening to identify patients at risk of developing cancer, screening for early detection

of cancer, determining prognosis, predicting response to therapy, and monitoring patients both during and after treatment. Molecular oncology testing has become the second largest segment of the molecular diagnostics market, as demonstrated by the large number of companies and tests that are discussed in Chapter 3. These include tests for breast, prostate, colorectal, and other solid tumors, as well as tests for leukemia and lymphoma. As one example, Genomic Health (Redwood City, CA) was a trailblazer with its *Oncotype DX* test for predicting breast cancer prognosis and response to chemotherapy, but several other competitors are entering this field. Personalized medicine has become an important part of molecular oncology testing, and several oncology tests to predict response have emerged.

The importance of personalized medicine is not limited to the field of oncology. Chapter 3 also includes a section on current and emerging examples of personalized medicine diagnostic tests that are being developed for other disease applications. These include tests for cytochrome P450, warfarin dose response, thiopurine methyltransferase (TPMT) genotyping, hypersensitivity to abacavir (due to the presence of the HLA-B*5701 allele), and others.

Another important segment of molecular diagnostics is genetic testing, which can be divided into two categories: testing for inherited disorders and testing for genetic changes that increase a person's risk of developing certain diseases. The markets for most tests for inherited disorders are small, although overall this is a significant segment due to the large number of inherited diseases that exist. One inherited disease that has emerged as a significant market opportunity for molecular diagnostic tests is cystic fibrosis, which results from a mutation in the cystic fibrosis transmembrane regulator (*CFTR*) gene. Other segments of the inherited disorders genetic testing market presented in this report include prenatal diagnosis, Ashkenazi Jewish genetic panel testing, comparative genomic hybridization arrays, and others.

An additional important aspect of genetic testing is predicting which individuals are at increased risk of developing certain disorders, based on their genetic makeup. Early applications in this field include tests offered by Myriad Genetics (Salt Lake City, UT) to predict the risk of developing breast cancer, colon cancer, or melanoma. Other (nononcology) applications of genetic testing to predict disease risk have also emerged. These include tests for predicting the risk of venous thromboembolism, cardiovascular disease, and others.

Chapter 4 discusses many of the business considerations associated with molecular diagnostics product development, commercialization, and marketing. This includes a discussion of the overall molecular diagnostics market, selected major trends in this market, the impact of personalized medicine and molecular diagnostics on pharmaceutical and molecular diagnostic companies, and selected major challenges facing molecular diagnostic companies.

Chapter 5 presents 10 expert interviews with executives at companies in the molecular diagnostics industry, including both companies that are developing and marketing kits that are sold to clinical laboratories and companies that have chosen the different strategy of operating their own CLIA-certified laboratories to offer testing services using their proprietary tests.

