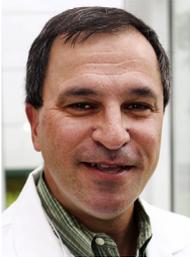


Preface

Current Topics in Molecular Diagnostics and Precision Medicine



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Editor

At the time of the writing of this editorial, the United States and for that matter the world were finally seeing the COVID-19 pandemic begin to dwindle to the lowest levels of SARS-CoV-2–driven mortality and decreases in new numbers of infections. Over the course of the past two years, laboratory testing became a household topic of discussion, yet most do not recognize that the vast majority of clinical decisions are made based on a lab test result. Nonetheless, the pandemic brought laboratory testing into the limelight, and the heroic efforts of laboratory staff everywhere were acknowledged by those who once saw the lab as a black box.

It has been almost 70 years since the first published description of the structure of DNA by Watson and Crick. No other section of laboratory medicine has experienced the explosive growth in molecular technologies and clinical applications as we have seen in molecular diagnostics. Beginning with relatively simple Southern blot transfer analyses and end-point polymerase chain reactions (PCR) through highly complex microarrays, digital PCR, and massively parallel sequencing, molecular technologies have impacted all facets of medical practice. Dramatic decreases in cost, vast improvements in turnaround times, and the ability to decentralize molecular technologies for use at point of care have resulted in unprecedented growth in implementation, reimbursement, and clinical utility.

As universal technologies, molecular techniques have become accepted as standard of care and routinely applied to the detection of nucleic acids for the identification of disease-causing variants, for the presence of pathogens, and to direct therapeutic selection as a means to inform precision medicine across the spectrum of human disease. From viral load testing where molecular disease monitoring made its debut to

current applications of liquid biopsy to assess cell-free DNA, the trend has been to provide complex information from the least-invasive sample possible. In addition, there is a huge need to democratize these technologies by developing rapid, user-friendly, low-cost mechanisms of testing for deployment outside of a central lab to point of care and at-home testing, which was so clearly evident during the pandemic.

The field of molecular diagnostics has matured. No longer do providers question the use of a molecular technique, they ask for it. Payors continue to improve reimbursement rates for those tests and technologies that offer the most cost-effective and clinically useful results, realizing that downstream savings in the management of a patient are real and significant. Federal regulators continue to review and approve testing for clinical use, and as we saw with the pandemic, recognized the need to allow labs to develop tests and follow guidance under the Emergency Use Authorization ruling. The public has become more engaged and educated in their health care needs/wants through the Internet and continue to participate in direct-to-consumer testing of various types. Together these attributes have made molecular diagnostic testing the standard of practice for many clinical diagnostic and management strategies.

In this special issue of *Clinics in Laboratory Medicine* titled “Current Topics in Molecular Diagnostics and Precision Medicine,” we explore the application of molecular technologies to a variety of human disease conditions. Articles by experts in their individual subspecialties highlight the utility of molecular technologies in understanding and detecting the cause of disease. I would like to thank the many contributors for sharing their thoughts in their excellent articles. There is no doubt that molecular diagnostics will continue to contribute to a better health care delivery system.

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