In the modern era, advances in clinical laboratory screening and diagnostic tests have had a profound impact on improving the quality and value of medical care (1, 2). Today, laboratory diagnostic tests can inform physicians and patients of the exact causes of a number of diseases and disorders, including cancer, chronic diseases, infectious diseases, sexually transmitted diseases, and genetic disorders. This precise ability to identify the nature of illnesses has led to developments of drugs and therapeutic methods that have saved millions of lives (3). Some diagnostic tests, such as those for glycated hemoglobin (type 2 diabetes) and BRCA1/2 (breast and ovarian cancers), are used for early diagnosis of at-risk patients, potentially preventing the onset of costly and deadly cancers and chronic diseases. Laboratory diagnostic tests have always served as the trailblazers of medical innovations by creating new pathways to identify and conquer difficult medical conditions.

A New Challenge for Laboratory Diagnostics in the 21st Century

Despite this impressive history of evolving diagnostic technologies that reshape and broaden our understanding of diseases, new challenges await us as we look ahead into the 21st century. Difficult-to-treat disorders such as Alzheimer disease, amyotrophic lateral sclerosis (Lou Gehrig disease), lupus, and multiple sclerosis are not well understood to be properly diagnosed and classified. Some chronically debilitating and deadly diseases such as cancers, migraines, and Parkinson disease can be diagnosed, but further research is needed to introduce better classification schemes, better preventive screening tests, and more quantitative tests that can effectively track disease progression.

Another challenge facing the clinical diagnostics community is the rising cost of health care. With U.S. direct health care expenses soon to reach 20% of annual gross domestic product, keeping health care costs under control is a national problem (4). So far, little effort has been put forth to make diagnostic tests more affordable and accessible. With genetic testing expected to become more prevalent in the future, the cost issues in laboratory diagnostic testing will only become more serious (5). Without a major paradigm shift, the confluence of these conflicting issues lowers the visibility on how research efforts, investment activities, and usage decisions will be made in the clinical diagnostics community.

What Is a Disruptive Innovation?

Technological innovations borne out of scientific discoveries have been the core drivers of all advances in fields of clinical diagnostics. For example, discoveries in optics gave rise to microscopes, which ushered in the era of laboratory diagnostics. The discovery of the genetic dogma paved the way for molecular diagnostics. These innovations have not only deepened our understanding of the diseases, but also transformed the way we provide medical care. Most of the new technologies, including some of the radical breakthroughs, improve the functions of existing diagnostic technologies along a performance trajectory. Early-stage technologies generally have limited functionalities, but sustain improvements over time. The early optical microscopes have given way to significantly more powerful electron and fluorescence microscopes, and these are being improved on today (6). The latest real-time PCR technology can complete some DNA amplifications in <60 s (7). Because these innovations make existing technologies better over time, they are defined as sustaining innovations.

Sometimes, new diagnostic technologies do not sustain the traditional trajectory of improvement. Instead, they offer solutions with different performance metrics or new value propositions (Fig. 1). Such diagnostic products tend to be more affordable and require less expertise to operate. Initially, these innovations might be limited in performance or designed more simply than existing technologies. Over time, however, they become more sophisticated and advanced, often equaling the performance and quality metrics of the incumbent solutions. These types of new technologies are defined as disruptive innovations, since they offer new paradigms in diagnostics (8).

Point-of-care diagnostic tests are good examples of disruptive innovations, because they broaden the accessi-
bility of the tests while making them more affordable. Although more validations on the technology are required, Theranos’s low-cost blood tests atWalgreens could substantially affect how basic diagnostic screening and tests are administered in the future (9).

Genetic tests marketed to the public are also potentially disruptive, because the tests are affordable and easily accessible. Another disruptive example can be found in genetic testing. Until last year, genetic tests offered by 23andMe were not approved by the US Food and Drug Administration (FDA) and were non-reimbursable, but with first FDA approval indicated in early 2015, these tests will increasingly gain recognition and acceptance by both medical and reimbursement communities (10).

Some sustaining innovations could also play a central role in the emergence of new disruptive diagnostic methods. The FDA cleared the mass spectrometry (MS)-based automated microbial identification system, VITEK-MS, in 2013 (11). Compared with traditional culture-based testing, the MS-based method requires much smaller testing samples, and the testing can be performed much sooner, reducing the total time required to complete the process. A rapid emergence of CLIA-laboratory based MS testing services will significantly reduce the time-dependent costs of diagnostic testing, potentially disrupting the current diagnostics landscape.

Why Do Disruptive Innovations Matter in Laboratory Diagnostics?

Sustaining innovations have enabled existing diagnostic technologies to be more sensitive, faster, and more powerful. We will need an even greater number of sustaining technologies to perfect many of the clinical diagnostics currently in use. Efforts and investments in translational research will also need to increase, to improve many of the nascent diagnostic technologies targeting difficult problems.

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2 Nonstandard abbreviations: FDA, US Food and Drug Administration; MS, mass spectrometry.
However, disruptive innovations in diagnostics may be where we could realize the biggest impact over the next several decades. Because all disruptive innovations ultimately lower costs of products without compromising their quality and performance, new disruptive diagnostic tests could significantly contribute to lowering of the costs of health care overall (12). Furthermore, integrating and leveraging already advanced technologies may hold answers for some of the most challenging diagnostic problems of today.

We look for the integration of gene sequencing and supercomputing, 2 sustaining innovations, to deliver key pieces to the puzzle for many hard-to-diagnose diseases. Wearable devices could push the frontier of point-of-care testing. Advances in rapid real-time PCR, MS, and other analytical technologies common in laboratory research could hold the answers to a new generation of molecular diagnostic tests that are cheaper, faster, and more accessible in the clinical setting.

Reaching the New Frontiers of Diagnostics via Disruptive Innovations

Because disruptive innovations introduce new products and services in health care in an affordable and accessible way, the time or investment required to introduce a new technology is well below that of sustaining innovations. The industry needs to embrace this opportune lower hurdle for developing disruptive diagnostics. Instead of waiting to perfect a technology, new diagnostic products need to be more quickly translated from basic science to the bedside, so that the iterations from real-world experience drive improvements and perfection.

The core of health care’s current landscape, in which the general hospital model still dominates, has not changed much for more than a century (13). This structure continues to dictate how patients are diagnosed, how results are interpreted, and who pays for tests. With the chronic disease population increasing along with the costs of care, the existing structure may not be well suited to address these changes in disease demographics. Similarly, the main objectives of laboratory diagnostics must be broadened to achieve not only an accurate diagnosis of illnesses, but also a delivery of lower-cost, more accessible screening and testing solutions. Disruptive innovations are the only solutions that fit these requirements concurrently. It is time to usher in new paradigms in clinical diagnosis.

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Author Contributions: All authors confirmed they have contributed to the intellectual content of this paper and have met the following 3 requirements: (a) significant contributions to the conception and design, acquisition of data, or analysis and interpretation of data; (b) drafting or revising the article for intellectual content; and (c) final approval of the published article.

Authors’ Disclosures or Potential Conflicts of Interest: No authors declared any potential conflicts of interest.

Acknowledgments: The author thanks the Clayton Christensen Institute for Disruptive Innovation for their support.