



# Transforming Healthcare with Next-Generation Genome Sequencing

## Policy Issues for Precision Medicine

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### EXECUTIVE SUMMARY

Next-generation sequencing (NGS) creates transformative opportunities to expand our understanding of biomedicine, reshape clinical care, and improve human health. NGS also opens broad vistas for innovation and economic growth.

DNA sequencing technology is among the most rapidly advancing technologies in history, and if the 19th century was the Age of Shipping and the 20th century the Age of Finance, the 21st century promises to be the Age of Genomics.

The magnitude and importance of these opportunities and the rapid pace of progress are leading many observers to call precision medicine the next global space race. These observers identify it as not only a revolution in healthcare, but also a strategic determinant of national competitiveness and prestige in the coming decades.

By embracing the fact that NGS is on its way to becoming universally available, we can accelerate progress toward that reality and prepare to take full advantage of it. We can position the United States to lead as a center of life sciences research, clinical and pharmaceutical discovery, and bioscience innovation.

With programs such as the National Cancer Moonshot<sup>1</sup> and Precision Medicine Initiative (PMI),<sup>2</sup> the United States is taking exciting steps toward precision medicine. But many other nations are also moving aggressively.

*Whole genome sequencing has the potential to add value for clinical and research applications, as well as for the health system as a whole. It can help shorten a patient's diagnostic journey, eliminate the need for repeated diagnostic procedures, and reduce reliance on one-size-fits-all treatments.*

Practical policies can help the United States advance its leadership in healthcare and clinical research, maximize the value of genomic data, and deliver the benefits of precision medicine to Americans. Recommended actions include:

- Continue federal funding for strategic health initiatives such as the Cancer Moonshot, Precision Medicine Initiative, and the Veterans' Administration's Million Veteran Program (MVP),<sup>3</sup> and ensure that genomics and NGS are essential parts of these programs
- Move toward collecting comprehensive genomic data via whole genome sequencing (WGS), which offers great power to drive exploration, discovery, and innovation
- Set guidelines that facilitate data sharing while protecting patient privacy and security
- Establish a standardized, machine-readable consent form for sharing genomic data, in alignment with standards work being done by the international genomics community
- Collaborate and advise in order to clarify and streamline the regulatory environment while ensuring efficacy, access, and patient safety
- Provide incentives for clinical genomics by establishing reimbursement guidelines and keeping clinical decision support (CDS) tools as part of Meaningful Use (i.e., the incentives specified for using certified electronic health records (EHRs) to achieve objectives defined by the Centers for Medicare & Medicaid Services (CMS)).<sup>4</sup>
- Create a glide path to flexible standards of care for precision medicine
- Facilitate the integration of genomic data into electronic health records and continue to advance EHR interoperability

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*If we're not collecting rich NGS data, we're losing a crucial resource for scientific exploration. Without this data, US leadership in precision medicine may be at risk.*

## The Added Value of Whole Genome Sequencing

In the past, the high costs and complexity of genome sequencing technology made it impractical to use whole genome sequencing routinely for most clinical and research purposes. Instead of capturing and assembling the billions of combinations for each location along the DNA strand, researchers looked only at targeted locations of the genome known to be relevant to the question at hand. These limited approaches include single nucleotide polymorphism (SNP) genotyping, which checks specific locations to test for a known genetic condition or ancestry information, as well as exome sequencing, which looks at the protein-encoding parts of the genome. If each human genome is a book, WGS "reads" the entire book. Narrower sequencing methods are akin to examining a few paragraphs, the table of contents, or even just a few letters at a given location in the "book."

While these limited approaches have value in specific situations, WGS has the potential to add value for clinical and research applications, as well as for the health system as a whole.

- **Increase diagnostic accuracy.** WGS has the potential to shorten the diagnostic journey of children with genetic diseases. In a study by the Garvan Institute of Medical Research's Kinghorn Centre for Clinical Genomics in Australia,<sup>8</sup> researchers found that WGS offered a significantly higher rate of confirmed diagnoses for rare monogenic diseases than whole exome sequencing (WES), the next most thorough examination of the genome.
- **Reduce healthcare costs.** By getting to the root of a problem faster, WGS can reduce the need for repeated tests and diagnostic procedures. This has the potential to increase the efficiency of the healthcare system, and may lower overall health costs by reducing the need to go back to the patient or research participant for additional sequencing if the initial level of sequencing proves insufficient.
- **Improve quality of life.** Establishing a diagnosis for patients with rare genetic disease—obtained quickly by noninvasive methods and followed by appropriate treatment—can reduce trial-and-error diagnosis and one-size-fits-all treatments. This can help reduce suffering and enhance patient and family satisfaction. By returning adult patients to health and productivity more quickly, it can provide an economic benefit, and when treatments are available, can give some children with rare diseases back their childhood.
- **Advance science and innovation.** While the number of targeted treatments is growing rapidly, genomic medicine is a field where much is still to be discovered. WGS gives researchers a powerful data set for exploration, insight, and eventual breakthroughs, which in turn may lead to new treatments, diagnostics, and preventive measures. The resulting innovations have the potential to improve health and healthcare and create economic growth.

## The Opportunity and the Vision

Next-generation sequencing combines sophisticated biomedical chemistry and imaging with advanced bio-informatics to index and analyze a genome. The techniques of NGS can be used to examine the 3 billion base pairs that comprise the whole human genome. The resulting data help identify disease factors and are proving to be invaluable in informing treatment decisions and prevention strategies.

Radical advances in NGS technologies have made genome sequencing faster, more accurate, and more affordable than ever (Figure 1). At the same time, Moore's Law and increasingly powerful analytics have advanced the bioinformatics that help make sense of the genetic information. In contrast to the first human genome, which was sequenced in a USD 3 billion, 13-year multinational collaboration, today's labs can sequence a whole human genome within several days at a cost of approximately USD 1,000.<sup>5</sup>

WGS provides the most comprehensive information on an individual's genome. (See sidebar, The Added Value of Whole Genome Sequencing.) Genome sequencing is reshaping medicine today, and with continued progress is expected to usher in an era of biomedical innovation and improved health.

Whether nations perform sequencing through an approach of "Every Baby at Birth," "Every Disease at Diagnosis," or "Every Insured Individual upon Enrollment," their clinicians and scientists gain powerful information that may help drive new methods to prevent, diagnose, and treat a wide range of diseases. In doing so, these efforts have the potential to advance the ability to deliver cost-effective healthcare services, advance reproductive health, and offer novel therapies while limiting harmful side effects. The nations that lead in the genomic space race have the opportunity to gain competitive advantages in generating new sources of economic activity and increasing national prestige and pride.<sup>7</sup>

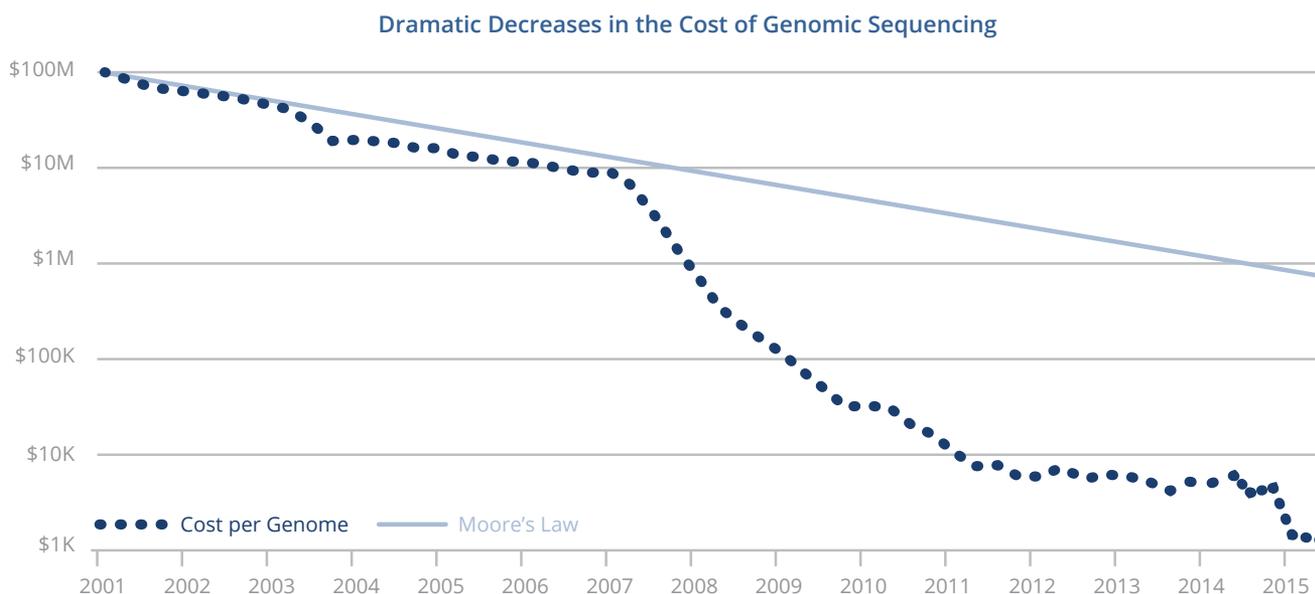


Figure 1. Dramatic decreases in the cost of genomic sequencing.<sup>6</sup>

## Progress Around the World

The United States is taking a number of positive steps that create a foundation for precision medicine. Whole genome sequencing should play a central role in these efforts. Since cancer is a disease of the genome, WGS can make valuable contributions to the National Cancer Moonshot, providing robust data for identifying factors leading to cancer formation and the possibility of inspiring new diagnostic methods and treatments.

The Applied Proteogenomics Organizational, Learning, and Outcomes (APOLLO) Consortium, announced in July 2016 by the Cancer Moonshot Task Force, illustrates the importance of both NGS and of interagency collaboration. The consortium, which includes the Department of Defense, Department of Veterans Affairs, and the National Cancer Institute (NCI), will combine NGS data with advanced methods for studying the actions of proteins in the body to gain insights that may lead to potential treatments.<sup>9</sup> WGS should also become an essential element of both the one-million-patient cohort now being developed as part of the Precision Medicine Initiative, and the Million Veteran Program being administered by the Veterans Administration.

In other positive actions, the US Food and Drug Administration (FDA) is moving to streamline the regulation of sequencing tests while ensuring safety and accountability.<sup>10</sup> The US House and Senate have focused attention on genomics in numerous hearings on issues related to biomedical innovation and healthcare, and we expect continued efforts to accelerate progress.

In the private sector, forward-looking healthcare systems recognize the importance of precision medicine, and are incorporating genome sequencing into programs aimed at advancing innovation and improving clinical care. For example:

- **Geisinger Health System**, a physician-led integrated system in Pennsylvania, has enrolled 100,000 individuals in its MyCode\* Community Health Initiative, in collaboration with Regeneron Pharmaceuticals.<sup>11</sup> The initiative, which plans to enroll 250,000 individuals, combines DNA sequencing with 20 years of EHR data, and provides both research and clinical benefits. Clinically relevant results are incorporated into the patient's EHR and used to inform clinical practice, while researchers are mining the data in search of new drug targets and therapies. Geisinger's Genomic Medicine Institute is using WGS to shorten the diagnostic journey of children and others with complex, undiagnosed conditions.<sup>12</sup>

*Performing WGS for every member of the Precision Medicine Initiative Cohort Program (PMI-CP) would provide an outstanding start on genomic insights, particularly since the PMI-CP is working hard to ensure that the cohort reflects the diversity of the nation's population.*

- **Intermountain Healthcare**, a not-for-profit system based in Salt Lake City, has established a Precision Genomics Core Laboratory that applies molecular analysis to patient care. In a clinical genomics collaboration, Intermountain will work with the Stanford University School of Medicine's Genome Technology Center to identify new biomarkers, conduct clinical population studies, and jointly develop innovative technologies.<sup>13</sup>

Globally, more than twenty countries are developing precision medicine initiatives, many based on WGS. For example, Genomics England, established in 2013 by the United Kingdom's Department of Health, is sequencing 100,000 whole genomes of National Health Service patients who suffer from cancer and rare genetic disorders.<sup>14</sup> The People's Republic of China (PRC) has announced a five-year, USD 3 billion precision medicine initiative that demonstrates its intent to lead in the field.<sup>15</sup> Current plans are to sequence at least one million individuals and incorporate genomics into a range of clinical and research applications. France is investing USD 745 million over five years to build the nation's genome sequencing capacity as a step toward personalized medicine.<sup>16</sup> Sequencing will occur at 12 centers, and by 2020 the country's leaders intend to be sequencing 235,000 samples per year for both clinical and research applications across a range of diseases.

## Roadblocks to Precision Medicine

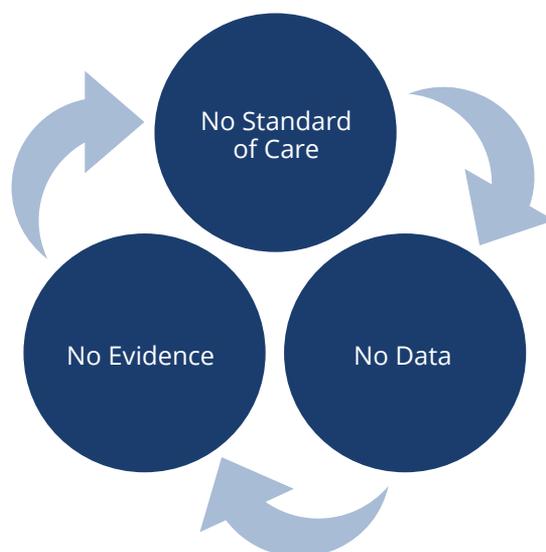
In a strategic field that is advancing rapidly, the progress of precision medicine in the United States is often slowed by issues arising from our distributed healthcare system, fragmented payer system, slow collection of genomic data, uncertain regulatory and policy environments, and others.

These issues impose difficulties on the work needed to advance clinical care and generate new research insights. They also contribute to a cycle of inaction identified in recent research by Intel ethnographers (Figure 2).<sup>17</sup> Focusing on cancer care, the researchers found that because routine NGS testing of all patients has not become the standard of care in the United States, oncologists are reluctant to order genomic tests unless they are confident the results will contribute to an actionable outcome—ideally a clear indication for an accepted treatment that will be covered by insurance or other funding. Payers are reluctant to cover the costs of NGS testing until they have clear evidence of the tests' benefit. But because oncologists aren't ordering the tests, the data to build a case for genomic testing as part of an evidence-based standard of care isn't being accumulated. And since NGS testing is not the standard of care for most cancers, payers feel justified in not covering it.

It's a vicious circle. And it is compounded by the need for education and expertise to interpret diagnostic results and counsel patients, and the lack of mature, trusted tools that incorporate genomic information into patients' health records and present it to clinicians at the point of care.

The results are serious. They include:

- **Lack of access to precision medicine.** Without NGS testing, patients who may benefit from new treatments or clinical trial opportunities based on genetic information, such as identification of a cancer's molecular variants, may not be offered those options.
- **Unequal access to precision medicine.** Because payers are not covering the costs of NGS testing, precision medicine is often available only to individuals who can pay the cost themselves, or who live near major urban medical centers that cover the costs as part of research initiatives. These inequalities are compounded by the underrepresentation of diverse groups in genomic data collection.<sup>18</sup> Because genetic mutations are expressed differently in different populations, this underrepresentation means that treatment recommendations and drug development may not accurately reflect the needs of all Americans.
- **Slowing of scientific advances.** Research breakthroughs can be expected to arise from collaborative efforts to combine and analyze disparate data sets, including genomic, clinical, lifestyle, pharmaceutical, environmental, and other information. If we're not collecting robust sequencing data, we're losing a crucial resource for scientific exploration. Without these data, US leadership in precision medicine may be at risk.



**Figure 2.** Cycle of inaction slowing adoption of precision medicine.

## Policies to Advance Progress

Policy modernization is crucial to breaking the cycle of inaction and advancing US leadership. Policies should focus on empowering the United States to advance science and innovation while protecting patients and ensuring that the benefits of precision medicine are widely and equitably available. As we develop standards for practical matters such as e-consent and data sharing, it can be beneficial to harmonize with international efforts—to capitalize on their work, contribute US expertise, and advance the ability to support scientific collaboration. Both Illumina and Intel are members of groups such as the Global Alliance for Genomics and Health (GA4GH).<sup>19</sup>

The following are suggested actions we can take to help align policies with evolving requirements:

### GATHER WGS DATA TO BUILD A SOLID FOUNDATION

Progress in genomic medicine starts from a base of population genomes. Because WGS provides more genetic information than other sequencing methods, it has great potential to drive exploration, discovery, innovation, and clinical breakthroughs. The more insights we gain into the genetic bases of health and illness, the greater our ability to develop precise treatment and prevention plans.

To build a solid foundation for innovation and clinical care, policymakers and legislators should make WGS an integral part of the nation's strategic health initiatives. Performing WGS for every member of the Precision Medicine Initiative Cohort Program (PMI-CP) would provide an outstanding start on genomic insights, particularly since the PMI-CP is working hard to ensure that the cohort reflects the diversity of the nation's population.

Policymakers should also ensure that appropriate programs encourage the use and sharing of sequencing data, as outlined in the Cancer Moonshot Blue Ribbon Panel Report.<sup>20</sup> This report laid out plans for “linking and leveraging a number of existing pilot programs across the nation that use next-generation genomic sequencing.” The report also acknowledged that “our ability to accelerate progress against cancer demands that researchers, clinicians, and patients across the country collaborate in sharing their collective data and knowledge about the disease.”

Other government-funded initiatives like the Veterans Administration's Million Veteran Program (MVP) should include provisions for sequencing and mandate coordination with other efforts that maximize existing investments and build a foundational data set that will be unrivaled. Although the data from MVP is a significant contribution, we encourage supplementing it by intentionally seeking data that represent more diverse populations, including women and varying ethnicities. To achieve the greatest clinical and research value, these programs should collect whole genome data.

## SHARE THE DATA TO ADVANCE RESEARCH AND PATIENT CARE

Research to develop precision medicine requires collaboration and data sharing among clinical institutions, pharmaceutical and medical device companies, researchers, life scientists, and technology innovators. Insights will come from combining genomic data with information from longitudinal clinical records, as well as with pharmaceutical, environmental, lifestyle, and other data. Researchers are eager to collaboratively analyze these diverse data sources in far larger quantities than any single clinical institution will possess, making data sharing a priority. Vice President Biden has emphasized the importance of data sharing to the success of the Cancer Moonshot.<sup>21</sup> The announcement that the National Institutes of Health will require grant recipients to share data is a major step to achieving access to the data.<sup>22</sup>

*Policies that encourage WGS testing can advance both the development of WGS standards of care and the science of precision medicine, which ultimately enlarges the universe of targeted treatments for genetically based illnesses.*

Policymakers can facilitate the collection and sharing of NGS/WGS data by making the appropriate sharing of genomic data an essential part of relevant, federally funded projects. Policymakers should support development of a standardized, machine-readable consent-for-sharing form that allows patients to donate their NGS/WGS data where and how they choose. In addition to using standardized consent language, the form should offer simple ways for participants to review and update their consent over time. While the greatest research value comes from patients' giving the broadest consent, this desire must be offset by respect for the patient's or citizen's autonomy. The PMI Cohort Program's goal of viewing citizens as partners in participatory research provides a promising approach to informed consent.<sup>23</sup>

Data sharing is also enhanced by having a standardized format for expressing genomic data. Harmonizing standards for capturing genomic data is crucial to providing semantic interoperability and allowing the data to be securely and efficiently integrated into patient health records and analytic software programs.

Policymakers should also continue their work to establish technical standards for interoperable electronic health records. Interoperable EHRs are important in enabling patients and their care teams to incorporate the results of genetic diagnostics into their clinical decision making. Allowing genomic and other clinical data and tools to interoperate in secure, trusted ways will generate a powerful network effect that amplifies the value of genomic data to researchers, healthcare professionals, and patients.

The ability to share large volumes of genomic and clinical data is enhanced by federated approaches to data storage. For example, the Collaborative Cancer Cloud,<sup>24</sup> which Intel and Oregon Health & Science University (OHSU) established in 2015, is an open-source analytics platform that enables medical institutions to securely share insights from their private patient genomic data for potentially lifesaving discoveries. The Cancer Cloud's federated storage enables large amounts of data from sites all around the world to be analyzed in a distributed way, while preserving the privacy and security of the patient data at each partner site. By using state-of-the-art technologies, the Cancer Cloud speeds genomic analysis and offers opportunities to enhance clinician productivity, giving physicians more time to work with patients and devise personalized treatments.

## PROTECT AND EDUCATE PATIENTS

To freely share their genomic data, patients must be confident that their privacy will be protected, their information will provide benefit to themselves or society, and they will not experience discrimination based on their genetic profile. This requires comprehensive policies, a thorough commitment to transparency, and programs to increase genomic literacy at all levels of society.

Strong privacy protections must include safeguards against the risks of patient identities being reconstructed from de-identified data. Policymakers and legislators must also ensure that genomic data will not be used for nonresearch purposes without explicit consent, and that data collected through federal programs will not be obtainable through Freedom of Information Act (FOIA) requests. Patients must not risk being unfairly denied health insurance or other program benefits because they've had their genomes sequenced. The Genetic Information Non-discrimination Act (GINA)<sup>25</sup> was an important step, but a 2014 Huntsman Cancer Institute Survey still indicated that, among respondents who would not seek genetic testing for cancer, 40 percent cited concerns it would impact employment and nearly 70 percent were concerned it would impact insurability.<sup>26</sup> This suggests a need for better education about the protections offered by GINA, a strengthening of the protection offered by GINA, or both. GINA protections should also be extended to military personnel, who have not been afforded GINA protections.

Transparency is crucial, particularly where there has been a historical basis for mistrust. Policies should forbid any collection, storage, or sharing of genomic data without the patient's knowledge and informed consent.

Patient education programs can help build trust and increase participation. Education programs can raise awareness of the benefits of genome sequencing and precision medicine, and give patients the confidence to act as empowered participants and advocates. Given the importance of diversity in data collection, policymakers should consider community-based outreach programs targeting traditionally underserved populations, and link these to specific steps to ensure equitable access to sequencing.

## EXPAND PAYMENT AND COVERAGE FOR CLINICAL GENOMICS

Providing a reimbursement path for NGS-based diagnostic testing is an important step to interrupt the cycle of inaction and remove an important barrier to precision medicine. Reimbursement policies for genetically based illnesses should encourage the use of sequencing for diagnostics, which has been found to improve diagnostic effectiveness.<sup>27</sup> Particularly if paired with efforts to facilitate data sharing, WGS diagnostics can also provide robust data to drive the development of new treatments and help identify existing treatments that can provide clinical benefits.

Reimbursement strategies should include evidence-based efforts to track outcomes and demonstrate clinical utility. These strategies should also assess benefits such as cost savings from improved resource utilization throughout the healthcare system. In addition, the American College of Medical Genetics and Genomics notes that the benefits of genetic diagnostics deliver a "profound value" to patients and family members, as well as driving clinical advances that can shape prevention and treatment.<sup>28</sup> Clinical utility will rise as the number of treatments targeting genetically based diseases expands, creating a virtuous cycle of research innovation and care improvements.

*To avoid delays in a rapidly evolving field, policymakers and regulators should establish guidelines for clinical decision support systems and their component elements based on their intended use and the seriousness of risk to the patient.*

### **INCREASE REGULATORY AGILITY AND ESTABLISH APPROPRIATE SCOPE FOR REGULATION FOR SAFETY AND EFFICACY**

Clinical use of precision medicine will benefit strongly from an agile regulatory environment that assures clinicians and patients of the reliability, safety, and validity of diagnostic test results while reflecting the rapid evolution of NGS technologies and applications. Since the proper scope of regulation in WGS and clinical decision support systems (CDS) presents novel issues with respect to safety and efficacy as well as rapid learning and iteration, it is essential that industry and regulators work together.

Clear, flexible guidelines on NGS diagnostic devices and laboratory-developed tests (LDTs) are necessary to spur investment and innovation in these tests and devices. Actions such as the FDA's 2016 draft guidance documents<sup>29</sup> are welcome steps toward a clearer, more streamlined environment. Continued actions will help advance economic growth, enhance our life sciences leadership, and improve public health. Close collaboration is a best practice, and should involve research organizations, healthcare leaders, patient advocacy groups, and private-sector innovators in the biomedical, technology, and medical device industries.

### **BUILD TRUST IN CLINICAL DECISION SUPPORT SYSTEMS**

The informatics side of precision medicine presents its own regulatory challenges. Data analytics systems will be able to crunch through vast quantities of data to develop new insights that, when validated, will feed into clinical decision support (CDS) systems. Incorporating these results into high-quality CDS systems is key to integrating precision medicine into patient care and enabling use by community oncologists and other physicians.

Clinicians and patients must have confidence that recommendations generated by these CDS systems are based on relevant, validated, and vetted data, and that the data has been analyzed appropriately. Yet data analytics algorithms are increasingly designed for continuous learning and continuous improvement. The static regulations that may apply to traditional medical device oversight will not serve patients and clinicians well in the face of these rapidly advancing CDS systems, and the lack of guidance on how they might be regulated creates a difficult environment for development and investment in these tools.

As with the regulation of medical devices and diagnostic tests, clinicians and the healthcare system will benefit from clear guidelines that emphasize patient safety and privacy while removing unnecessary obstacles to innovation. To avoid delays in such a rapidly evolving field, policymakers and regulators should work with industry to establish guidelines for CDS systems and their component elements based on their intended use and the seriousness of risk to the patient.

Establishing alternative approaches to adverse-event reporting when appropriate and using consensus standards reflecting the efficacy of the decision support algorithms can also help to bring precision medicine's benefits to patients while ensuring patient safety.<sup>30</sup> For example, analytic validity and clinical validity should be separately reviewed. Risks attributable to a defect in the analytical validity of a software product can be sufficiently mitigated through compliance with industry consensus standards for software quality that ensure mathematically accurate calculations at a high degree of confidence. Likewise, because the clinical validity of an algorithm can typically be demonstrated through references to peer-reviewed literature, clinical practice guidelines, data registries, and other sources, FDA should not ordinarily require manufacturers to conduct new clinical trials to demonstrate clinical safety and efficacy.

## ADDRESS PRACTICAL ISSUES FOR CLINICIANS

In addition to robust CDS tools, clinicians need incentives, guidelines, and security if they are to break the cycle of inaction and deliver the benefits of precision medicine. Policymakers should ensure that CDS remains an integral element of Meaningful Use requirements and provides strong incentives for adoption. In addition, they should collaborate with clinical and biomedical leaders to develop a glide path that will result in establishing standards of care for precision medicine and WGS.

Standards of care can give clinicians leeway to explore and adopt precision medicine while minimizing risk. For example, if a patient's genome has been sequenced, how should the clinician proceed in the event of presymptomatic conditions? Are clinicians at risk if they collect WGS data but do not act aggressively on every potential problem? If they do act based on the best understanding at the time, but that knowledge changes with subsequent data collection and analysis, how are clinicians assessed?

Flexible standards of care can guide clinicians and provide malpractice protection if their clinical judgment deviates from the current, one-size-fits-all standard of care, including an evidence-based decision to avoid the established standard of care when precision medicine analytics show that the standard treatment is not likely to be effective for this particular patient. Liability protection should be offered to encourage clinicians and institutions to track patient outcomes, share early findings, and advance knowledge.

Encouraging WGS testing can advance both the development of WGS standards of care and the science of precision medicine, which ultimately enlarges the universe of targeted treatments for genetically based illnesses.

## EXPAND THE LIFE SCIENCES WORKFORCE

Integrating precision medicine into clinical workflows calls for educating physicians, expanding the nation's life sciences workforce, and building skills to develop and utilize the advancing capabilities.

Policies should support the development and accreditation of curriculum and programs for new medical professionals, along with continuing education for practicing physicians. Education should also address the growing need for genetic counselors and specialists to review test results and help oncologists and community physicians develop customized treatment plans.

Initiatives such as the education outreach program from the HudsonAlpha Institute for Biotechnology provide a useful model for promoting genetic literacy and inspiring a new generation of life scientists. HudsonAlpha educated more than 700,000 students and teachers last year through hands-on classroom modules, digital learning experiences, curricular materials, and in-depth school and summer camp experiences.<sup>31</sup>

Skills programs are needed to build a life sciences workforce that can support and advance the emerging capabilities. In addition to basic and applied scientists and researchers, workforce development should target people who will fill jobs in laboratory operations, data center management, data sciences, computer security, data storage, software development for life sciences applications, and others.

## BUILD THE INFRASTRUCTURE

Computation and informatics play crucial roles in genome sequencing itself, as well as in storing and analyzing the resulting data. Researchers will have the potential to drive clinical improvements by combining this data with clinical, pharmaceutical, and other databases. Precision medicine will also build on data from the emerging world of smart, connected “things,” ranging from state-of-the-art lab instruments to sensor-based devices that monitor patient health. Using increasingly sophisticated cognitive computing capabilities to analyze these vast data stores, life science researchers and clinicians expect to usher in a new era of computational biology.<sup>32</sup> The result may one day give clinicians sophisticated modeling and simulation capabilities that will enable researchers to model and predict a disease's progress.

Investments in infrastructure for precision medicine should be based on open platforms and industry standards, using technologies that are highly scalable, energy-efficient, and interoperable. This open standards-based approach has the potential to reduce costs and facilitate collaboration at all scales, from the supercomputer center to the clinician's tablet or wearable or even implantable health devices.

Programs such as the US Department of Energy's National Strategic Computing Initiative (NSCI),<sup>33</sup> which are advancing the development of extreme-scale computers, will be key long-term enablers for precision medicine. Computing architectures for precision medicine must support both analytics and simulation workloads, putting a premium on massive data storage capabilities and high throughput. NSCI's focus on multi-agency collaboration, industry partnerships, and scalable architectures represents an effective approach to meeting the nation's future needs for computational capacity for precision medicine and other strategic fields.

Next-generation 5G networks will be foundational for providing unprecedented intelligence and flexibility across communication networks, enabling the aggregation of data from billions of smart, connected health devices and other sensor-based elements. The speed of 5G networks will also support collaboration to combine and analyze the data with other types of information, and apply the results to drive new insights. The Federal Communication Commission's opening of the high-band spectrum for 5G networks is a positive step to harnessing the speed of these networks for precision medicine.

To ensure that next-generation platforms meet precision medicine's requirements, policymakers should also continue efforts to bring together Department of Energy's computational scientists and life science researchers and clinicians at agencies such as the Department of Health and Human Services and the National Institutes of Health. For broader issues, effective collaboration can also include regulatory agencies and representatives from the technology, pharmaceutical, healthcare, health instruments, research, and academic sectors.

## Conclusion: A Cycle of Innovation and Improving Health

As we begin to unlock the secrets of the genome, nations around the world are competing to strengthen their scientific leadership and gain the benefits of precision medicine. Policy modernization and participation in international standardization efforts are crucial to keeping the United States competitive in this strategic arena and ensuring that precision medicine's benefits are shared equitably by all Americans. Through comprehensive efforts to increase agility in the regulatory environment, advance the collection of WGS data, and facilitate the use of precision medicine, policymakers have opportunities to spur a cycle of biomedical innovation that will improve health, transform healthcare, advance the nation's life sciences leadership, and boost the US economy.

Illumina and Intel are at the forefront in enabling the Age of Genomics. Illumina is committed to unlocking the power of the genome, and the company and its customers are leading the application of next-generation sequencing to advance precision medicine and solve some of the world's toughest problems. As of 2015, Illumina technology was generating as much as 90 percent of the world's sequencing data.<sup>34</sup> Intel powers much of the world's bioinformatics and data analytics infrastructure—from wearable devices that collect real-time evidence via sensors, to clinical tools at the point of care, to the world's most massive supercomputers. Intel is committed to a vision of enabling patients to have their DNA sequenced and receive a precision treatment plan based on their unique biomolecular profile, all within 24 hours. Intel calls this vision “All in One Day,” and is working with stakeholders across the technology and biomedical industries to make it a reality. Both Illumina and Intel share expertise with policymakers, bioscience researchers, health innovators, and others to collaboratively deliver the promise of precision medicine.

## Learn More

Learn more about Intel in the life sciences and the All in One Day initiative. Talk to your Intel representative, or visit [www.intel.com/content/www/us/en/healthcare-it/life-sciences.html](http://www.intel.com/content/www/us/en/healthcare-it/life-sciences.html) or [www.intel.com/healthcare/bigdata](http://www.intel.com/healthcare/bigdata)

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