Accessing Genomic Medicine: Affordability, Diffusion, and Disparities

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Advances in the understanding of the genome, combined with affordable sequencing techniques and innovations in drug development, are ushering in an era of molecular diagnostics and individualized therapeutics with the potential to meaningfully enhance human health through effective prevention, diagnosis, and treatment. As the genetic origins of many diseases become better understood, risks and prognoses can be assessed based upon knowledge of genetic mutations, while biomarkers are becoming available to predict pharmacologic response in unique patients. This revolution in genomic medicine, sometimes also called personalized or precision medicine, has the ability to advance both disease prevention and patient outcomes.

These tools emerge into a health care system that has 5 characteristics that will shape the accessibility, management, and financing for these new technologies. In addition, questions of value, clinical management, and patient engagement will influence the emergence of personalized medicine.

First, increasingly unaffordable delivery systems are forcing societies across the world to ask fundamental questions about value. In the United States, by 2021, health care spending is estimated to be one-fifth of the economy with per capita spending increases between 5% and 6% a year.1 Advances in medical technology are a major contributor to these increases in health care spending, accounting for roughly half of the increase.2 As with many new technologies, genetic and genomic innovations will contribute to those cost pressures. For instance, at UnitedHealthcare, the annual average increase in the use of genetic and molecular diagnostics was about 14% from 2008 to 2010.3 Furthermore, a significant escalation in biologically derived specialty drugs accounted for nearly 20% of annual per-member pharmacy spending in 2011 and is expected to reach 25% of total annual per-member pharmacy spending by 2014.4

An encouraging development is that molecular advances, such as companion diagnostic tests that are linked to a specific drug or therapy, have the potential to decrease health spending. Personalized care approaches will replace expensive therapies that may be ineffective in individuals with a certain genetic and biological make-up. However, use of new personalized care has the potential for unnecessary utilization, with associated costs and potential for harm.5 While it is difficult to identify how the balance of new costs and savings will be resolved, it is clear these innovations will change current care delivery environments and their use must be responsibly managed. This is true for developed economies and for countries struggling with both infectious and chronic diseases.

Second, the Institute of Medicine recently estimated that the health care system wastes or misuses about $750 billion annually, in part due to failure to comply with evidence-based guidance, lack of timely information, inadequate incentives, and administrative oversight.6 Without substantial intervention, the increasing complexity of decisions associated with molecular medicine will exacerbate this problem.

Third, heritable disorders, alone or with complicating environmental factors, often produce chronic illnesses that require access to care across multiple settings and from multiple health and social service providers. However, care delivery today is too often fragmented and uncoordinated. Individuals who have chronic conditions with genetic origin may benefit from diagnostic and therapeutic advances but will simultaneously exert more pressure on already challenged systems, especially in underresourced communities.

Fourth, disparities in health outcomes resulting in part from differential access to disease prevention support and quality clinical care have been a persistent challenge for racial, cultural, and socioeconomically disadvantaged populations. Additionally, genomic information demonstrates that many serious diseases differentially affect subpopulations. The cost and complexity associated with genomic medicine have the potential for exacerbating disparities absent significant attentiveness to ensuring that technologies intended for clinical use are both effective and affordable.

Fifth, payer and purchaser demands to achieve greater value have stimulated new delivery models that respond to reimbursement alignment with demonstrated quality and cost-effectiveness benefit to patients. Patient-centered medi-

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cal homes and accountable care organizations will intensify management and measurement of new technology application.

If molecular-based innovations are to be effectively and equitably introduced into delivery systems, all stakeholders will need to work diligently to address the following issues.

Value Demonstration

New genomic-based interventions must be accompanied by convincing evidence, and standardized criteria, to support and align stakeholder evaluation of their value. Enhanced infrastructure for comparative effectiveness research in genomics should be a higher priority.

Key to value determination is proof of the accuracy and reliability of genetic tests. Despite more than 12 years of concerns and recommendations from prominent advisory committees, oversight remains suboptimal. More effective coordination is necessary from governmental and nongovernmental agencies involved in ensuring that genetic tests demonstrate both analytic and clinical validity before being introduced into the care system.

Clinical utility, another fundamental determinant of value, requires demonstration that innovations provide greater value than existing interventions in actual practice. Interoperable electronic health records and data exchanges must facilitate these evaluations. The National Institutes of Health’s genetic testing registry is a welcome addition deserving support.

The deficiencies of the medical coding system for molecular tests need to be addressed. The majority of these tests have no identifiable codes and, therefore, no way to identify why a test was ordered or its effect on clinical care. Priority attention should be given to enhancing the number and specificity of codes for describing genomic tests, developing an alternative genomic test coding system—the creative adaptation of code sets (such as the International Classification of Diseases, Tenth Revision diagnosis codes), or both.

Clinical Management

The complexity of the molecular era requires enhanced support for physicians to translate scientific discovery into clinical guidance. Additionally, because the number of facts associated with genomic clinical decisions meaningfully challenges human cognitive capacities, professional development must be coupled with electronic, evidence-based, point-of-care information. Given trends in payment policies that reward hospitals and physicians for quality and outcomes (performance-based reimbursement), priority should be given to the translation of clinical guidance into the measures upon which reimbursement will be based.

Patient Engagement

Genomic medicine will increasingly present patients and health professionals with complex and sensitive choices. Thoughtful engagement of patients in shared decision making requires attentiveness to genetic literacy and informed consent within the context of cultures. In short supply, genetic counseling service demand will require increased training support and efficient integration into care delivery teams, including telemedicine facilitation. People must feel comfortable in exploring their genetic risks for disease, which makes patient information security an inanimate priority everywhere.

Conclusions

Medical innovation that enhances human survival and relieves disease burden has been a capstone of the healing sciences and continues to drive advances in genetics and genomics. World realities, however, demand focus on the appropriate use of genomic medicine if all are to experience affordable, safe, and equitable access to their benefits. Health care stakeholders from the “bench to the bedside” have critical roles to play. While the challenges are significant, the opportunities are even greater.

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