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NEWS AND COMMENTARY

Genomic Medicine

'Grand challenges' in the translation of genomics to human health

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Despite clear advances in technology that bring genomic information closer to physicians, patients, and the public, looming even closer are issues that are outside the sphere of the genome sciences and more in the realm of genome policy. Recently, Scheuner *et al*[1] carried out an extensive meta-analysis of 68 studies seeking to integrate genomic medicine to the clinical management of chronic diseases. The study posed the following questions. What are the outcomes of genomic medicine? What is the current level of consumer understanding about genomic medicine. What information do consumers need before they seek services? How is genomic medicine best delivered? What are the challenges and barriers to integrating genomic medicine into clinical practice? The authors conclude that gaps in provider and public education, addressing privacy concerns, building an evidence base, and developing the appropriate cost models for genomic medicine in health-care delivery will need to be filled in order to facilitate the translation of genomic innovations into clinical practice. These findings are not surprising given the spectrum of genome policy issues that have emerged as the genome sciences have matured.[2] Nonetheless, this is the first comprehensive analysis of data on these issues and provides a good basis to address them.

Health professional and public education

The primary-care workforce feels woefully unprepared to integrate genomics into regular practice.[1] Consumers are enthusiastic about genetics and are hopeful about its impact, but at the same time they have a low knowledge base of both genetics and genetic testing for common diseases. Education of health professionals and the public must be a priority to advance the use of genomics into healthcare.[3] With the rapid advances in genomics research and developing technologies, it will be challenging to keep health professionals informed about the benefits, risks, and limitations of new tools as they become available. In addition, the public and health-care workforce will need to understand the appropriate clinical applications of genomic tools – including their benefits, risks, and limitations – and how they may improve clinical management. Direct to consumer genomic testing has only served to greatly intensify the educational needs and have made them more urgent across the genomic medicine community, from the common public to health-care providers to policy makers.[3] Several surveys have documented the below-average physician knowledge of genetics,[4] but none has assessed knowledge of the newer field of genomics. The importance of education in the application of pharmacogenetics has been described,[5] but at present there are no broad initiatives to orchestrate genetics and genomics education of medical professionals, trainees, and the public at large. Basic genomic literacy is a critical need for patients, physicians, and communities to engage in genomic research, and clinical studies are required to bring about a change in the care paradigms to support clinical genomics applications.

Privacy fears

Consumers are worried about the possible adverse consequences of genetic testing, particularly the privacy issues and discrimination against receiving employment and health insurance.[1] These concerns are coupled to angst regarding the lack of regulatory oversight of genetic testing. The uniqueness of genomic information is clearly debatable over whether it warrants special protections beyond those in place for standard medical information.[2] In the United States of America, fear of discrimination by employers and health insurers is the main concern, whereas in the United Kingdom, use of genetic information by life insurers is the major concern.[6,7] Regardless, the attention paid to genetics by the popular press and public has raised genetic information to a different level compared to other medical information. In order for genomic medicine to be integrated into routine clinical practice, associated fears with this type of testing must be put to rest. In the United States of America, many states have enacted legislation to protect against genetic discrimination by employers and health insurers; and in April 2008, the US Congress passed the Genetic Information Non-discrimination Act that will afford national protection of this information – a clear step forward in lowering the barrier to both research participation and clinical use of genomic testing.

Building the evidence for clinical utility

The most important factor hindering the appropriate integration of genomics into clinical practice today is the lack of evidence for its clinical utility (ie, evidence that use of a genomic

technology improves health outcomes). A paucity of randomized controlled trials has been performed to date examining clinical, behavioral, or psychological outcomes.[1] Practical issues for these types of studies need to be resolved, which will require greater collaboration among stakeholder groups and innovation in both study design and analysis methods. The design features of these trials must link the genomic test to outcomes via specific, actionable clinical recommendations. Financial support for these studies is often lacking as there are few incentives for manufacturers of genetic and genomic tests to make this investment. Public–private partnerships will likely be required to generate the evidence base for genomic medicine. These collaborations are desirable because firstly, no single stakeholder group is likely to have sufficient resources or expertise to conduct the necessary studies, and secondly, both will likely benefit from their execution.

Cost

Cost uncertainty (both in terms of delivery and reimbursement for genomic testing) is an important issue to many of the genomic medicine stakeholders.[1] As with any new innovation, genomic testing must be demonstrated to be clinically useful, cost-effective, and of value. But because genomic technologies inherently involve diagnostic or prognostic testing, and the complexities of incomplete gene penetrance and multiple gene and environmental interactions, their assessment can be more challenging. In addition, perhaps more than in any other area of medicine, questions have arisen concerning the economic incentives to develop these technologies. Formal health economics frameworks can be used to gain insights into these issues, and provide guidance for research and development investment, technology appraisal, and policy development.[8,9] Clarity is needed on the drivers of cost-effectiveness of genomic technologies and consideration must be given to approaches that include value-based reimbursement for genomic testing technologies.

The need for an integrated approach

The gaps to be filled to bring genomic medicine to fruition are exceedingly complex. In the United States of America, the Institute of Medicine's Roundtable on Translating Genomics to Health and the Centers for Disease Control's Evaluation of Genomic Applications in Practice and Prevention have independently developed programs aimed at developing a clear understanding of pathways for translation, the barriers that lie in the translational path, and the strategies to overcome them. It is clear that a broad stakeholder community of researchers, clinicians, patients, the public, industry, and policy makers must be convened to facilitate the kind of dialogue, exchange of information, science, and health policy planning that will be required to break down the barriers Scheuner *et al*[1] have described, and advance the genomic sciences to its most important end point of improving human health■

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