

Genomics in Health Care

Call for Papers for the 2013 Theme Issue

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SINCE THE 2001 PUBLICATION OF THE DRAFT HUMAN genome sequence,¹ genomic technologies have revolutionized the conduct of biomedical research. Current studies, many of which would have been unimaginable as few as 5 years ago, are reshaping understanding of the foundations of health and disease in profound and often unexpected ways. Most important to the practicing clinician, advances in genomic technologies are yielding tangible benefits to increasing numbers of patients. Although some have bemoaned the relatively slow pace of translation of genomic discovery to improved health outcomes, it is important to recognize that the intersection of genomic science and health care is in its infancy.²

Despite this, today, virtually every medical specialty is affected by genomic advances, although the influence of these advances in day-to-day care varies. For example, in a report recently published by a major health insurer, molecular diagnostics were estimated to account for 8% of expenditures on laboratory diagnostic testing in the United States in 2010, with projected double-digit yearly growth rates through 2020.³ In the setting of limited health care resources, the rapidly expanding expenditure on molecular diagnostics affects even those clinicians who have never ordered a genetic test. The advent of very low-cost approaches for genome sequencing promises to further accelerate the discovery process, and for individuals affected by currently untreatable conditions the advances cannot come too quickly.

Considerable challenge and opportunity exist at the intersection of genomics and health care. Five topic areas spanning the continuum of translational research stand out. First, human genome structure and function are dauntingly complex. Much remains to be learned about gene structure, function, and regulation and how these processes relate to normal and disease phenotypes in the context of differing environments. New experimental and computational approaches are needed to sort out this complexity.

Second, assigning causality (or at least a probability of causality) to newly detected variations in an individual's genome is currently an extremely arduous and costly task. This limits the ability to harness the potential of

genome sequence information for both clinical research and care. Without novel methods for providing near real-time, low-cost interpretation of the genome, use of high-throughput technologies like next-generation sequencing will be limited to a subset of highly resourced academic medical centers.

Third, robust and cost-effective strategies need to be developed for generating evidence of benefit for emerging genomic technologies. Genomics is coming of age during a time of global fiscal budget austerity, and traditional clinical trials are extremely expensive. Models for funding clinical trials that have worked for blockbuster drugs are not likely to work for molecular diagnostics and other genomic technologies with small target populations or low profit margins. Lack of an evidence base, measured in terms of improved morbidity and mortality, will hinder clinical guideline development and the willingness of payers to reimburse for genomic technologies.

Fourth, relatively little is known about how to deliver genomic services to ensure equitable, effective, and efficient care. Ample evidence suggests that there are significant knowledge deficits among clinicians and the public regarding genomics, and there is a shortage of health care professionals adequately trained to deliver genomic services.⁴ This suggests that proven genomic technologies must be integrated into health care workflows in ways that are not dependent on clinician expertise. In all probability this will necessitate development of electronic clinical decision support tools that are capable of providing point-of-care resources for consumers and health care professionals.

Fifth, genomic technologies are challenging the boundaries of what is possible vs what is desirable in the context of human health and health care. Direct-to-consumer genetic testing, the blurring of lines between clinical care and research, and assisted reproductive technologies are but a few of the arenas in which questions about genomics and society have rapidly transitioned from theoretical to concrete. Proven frameworks for education, communication, and decision making are needed to guide health care consumers, clinicians, health care organizations, and policy makers in the current time of often highly polarized health care discourse.

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To focus attention on and inform readers about these important issues, in April 2013, *JAMA* will publish a theme issue and The JAMA Network Journals⁵ will publish theme issues or articles devoted to genomics in health care. We are pleased to announce a call for submissions of original scholarly works related to this topic. For the purpose of the issue, *genomics* will be liberally interpreted to encompass investigations involving a holistic approach to understanding how genes and gene products interact with each other and the environment to result in health or disease. Manuscripts addressing 3 major thematic areas related to genomics will be considered for publication: (1) advances in the understanding of health and disease states; (2) advances in prevention, diagnosis, and treatment of health conditions; and (3) advances in the translation of genomics to improved health outcomes in individuals and populations, including issues related to a broader context of genomics in society. In addition to original research reports, we also invite submission of authoritative review articles and scholarly Viewpoints that inform the topic of genomics in health care.

Authors are encouraged to consult the Instructions for Authors for guidelines on manuscript preparation and submission.⁶ Importantly, articles of high scientific qual-

ity that are not of sufficiently broad interest for *JAMA* may automatically be considered for publication in appropriate subspecialty journals in The JAMA Network. Manuscripts received before November 1, 2012, will have the best chance for consideration for inclusion in the 2013 theme issue on Genomics in Health Care. The editors look forward to receiving your manuscripts for this important issue of *JAMA*.

Conflict of Interest Disclosures: The authors have completed and submitted the ICMJE Form for Disclosure of Potential Conflicts of Interest and none were reported.

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