FEATURED NEW INVESTIGATOR

Stakeholder consultation insights on the future of genomics at the clinical-public health interface

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In summer 2011, the Centers for Disease Control and Prevention Office of Public Health Genomics conducted a stakeholder consultation, administered by the University of Michigan Center for Public Health and Community Genomics, and Genetic Alliance, to recommend priorities for public health genomics from 2012 through 2017. Sixty-two responses from health professionals, administrators, and members of the public were pooled with 2 sets of key informant interviews and 3 discussion groups. *NVivo 9* and manual methods were used to organize themes. This review offers an interim analysis of progress with respect to the final recommendations, which demonstrated a strong interest in moving genomic discoveries toward implementation and comparative effectiveness (T3/T4) translational research. A translational research continuum exists with familial breast and ovarian cancer at one end and prostate cancer at the other. Cascade screening for inherited arrhythmia syndromes and hypercholesterolemia lags stakeholder recommendations in the United States but not in Europe; implementation of health service-based screening for Lynch syndrome, and integration into electronic health information systems, is on pace with the recommended timeline. A number of options exist to address deficits in the funding of translational research, particularly for oncogenomic gene expression profiling. The goal of personalized risk assessment necessitates both research progress (eg, in whole genome sequencing, as well as provider education in the differentiation of low- vs high-risk status. The public health approach supports an emphasis on genetic test validation while endorsing clinical translation research inclusion of an environmental and population-based perspective. (Translational Research 2014;163:466–477)

Abbreviations: CDC = Centers for Disease Control and Prevention; CF = cystic fibrosis; CTSA = Clinical and Translational Science Award; EGAPP = Evaluation of Genomic Applications in Practice and Prevention; FHH = family health history; GEP = gene expression profiling; NIH = National Institutes of Health; OPHG = Office of Public Health Genomics; PR = Priorities Report; RFI = Request for Information; SSRI = selective serotonin reuptake inhibitor; UHC = United Healthcare; WGS = whole genome sequencing

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Submitted for publication November 19, 2013; revision submitted December 18, 2013; accepted for publication December 19, 2013.

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1931-5244/$ - see front matter

http://dx.doi.org/10.1016/j.trsl.2013.12.007

Professionals attending the midwestern annual meeting of the Central Society for Clinical and Translational Research (CSCR) and the American Federation for Medical Research (AFMR) are treated yearly to an eye-catching view from above. From where the meeting takes place, city skyscrapers and residential lowlands can both be spotted, together with the numerous highways where they lead. Health professionals incorporating genetics into their arsenal of diagnostics and predictive testing can likewise benefit from a pervasive view of where genetic testing may lead their field.

In summer 2011 the Centers for Disease Control and Prevention (CDC) Office of Public Health Genomics (OPHG) conducted a stakeholder consultation, administered by the University of Michigan Center for Public Health and Community Genomics, and Genetic Alliance, to recommend priorities for advancing the field of public health genomics from 2012 through 2017 ("Priorities Report" [PR]).[1] The consultation generated recommendations that bear directly on the translation of genomic findings for the clinical community and highlight the value of a public health genomics perspective for ongoing movements in the medical field. The recommendations touch on key areas such as evidence-based medicine, translation of benchside research into useable guidelines and tools for practice, and absorption of genetic data into electronic information systems.

Of special importance is the "value added" benefit the consultation offers to clinical evolution. The public health perspective is, of necessity, population based. Although it is not to be expected that many forms of genetic testing will transcend the clinical setting, as they make their way into mainstream use, they will be offered to increasing numbers of individuals being managed in primary through tertiary healthcare centers. Public health also incorporates an appreciation of the environment's effect, which has a bearing on the interpretation of genetic tests. Lastly, public health has much to say about personalized risk assessment, both in regard to its support through translational research and coverage of the family as well as the individual.

In this review we offer an interim analysis of those recommendations straddling both the clinical and public health domains, considering where genomics has moved since the public consultation took place and suggesting areas deserving greater attention than what is currently being offered.

CONSULTATION PROCESS

Between June 30 and August 1, 2011, a CDC-OPHG Federal Registrar Request for Information (RFI) on the future of public health genomics drew 62 responses from a broad array of health professionals, medical and public health administrators, and public advocacy. The principle question asked was: What are the most important activities that should be carried out by the public health system in 2012–2017 to apply genomic knowledge to public health goals? Additional questions dealt with outcomes to be achieved; policies needed; institutions, organizations, and agencies to be involved; and potential barriers and solutions for incorporating genetics. A planning committee composed of genetics and health-related professionals—both practice and academe—and a representative from the Prevention Research Centers-associated National Community Committee convened to review the RFI responses and other inputs, and to refine models for organizing the data.

An initial list of major topic areas and subthemes was identified from the existing literature (59 articles + 2 books) and federal health agency and commission reports involving expert and public consultation on genetic technologies. *NVivo9* and manual methods were used to organize the RFI responses into tabular form. RFI responses were pooled with the results of 9 interviews of key informants from public health practice, academe, and the community; and 8 interviews of key informants from the healthcare for-profit and nonprofit sectors to yield a list of preliminary recommendations. Formatting of the recommendations was based on Institute of Medicine-developed public health core functions,[2] the *Public Health in America* essential public health services,[3] and planning committee-identified major themes (PR, pp. 14–5).[1] Input was then received from 3 discussion groups at Genetic Alliance's annual meeting, adding voices from the community, public health practitioners, and genetic counselors. The CDC-OPHG hosted a culminating day-long meeting of diverse stakeholders to solidify final recommendations and verify metrics (Table I).[1]

Clinical and public health genomics exist along a translational research continuum that moves from basic discovery aimed at candidate health applications (genetic testing and interventions) (T1) and assessment of readiness for health practice using evidence-based guidelines (T2); to delivery, dissemination, and diffusion research (T3); and research evaluating outcomes in the field through surveillance and comparative effectiveness analysis (T4) (PR, pp. 73–6).[1,4] Stakeholders lending their voice to the genomics consultation demonstrated more commonality than division regarding what should be taking place in the genomics arena in the intermediate future. This common territory, linking public health insights with clinical research and practice, will be explored in the pages to follow.