Short Communication

Public Health Genomics

Public Health Genomics 2010;13:360–361
DOI: 10.1159/000262327

Received: July 22, 2009
Accepted after revision: August 2, 2009
Published online: November 26, 2009

A Genetic Test Registry: Bringing Credible and Actionable Data Together

M.J. Khoury[a] M. Reyes[a] M. Gwinn[a] W.G. Feero[b]

[a]Office of Public Health Genomics, Centers for Disease Control and Prevention, Atlanta, Ga.,
[b]National Human Genome Research Institute, National Institutes of Health, Bethesda, Md.,USA

Javitt et al. [1] propose a blueprint for a mandated genetic test registry to support informed health care decision making. According to the authors, the core aim of the registry is 'to promote transparency, which includes disclosure of both what is known and what is not known.' They propose that for each test, the registry should include not only a description of the test and its intended use, but also information about its analytic and clinical validity, clinical utility, and proficiency testing.

We agree that reporting data on validity and utility is an essential element of quality assurance in genetic testing. Nevertheless, whether reporting to a genetic test registry is mandated or not, there is a fundamental challenge in implementation of such a registry, particularly with regard to the sources, standards and quality of submitted data and their systematic evaluation. As acknowledged by the authors, laboratories performing genetic tests may have only some of the data needed to evaluate validity and utility of tests. In particular, information on clinical validity and utility will have to be brought together from multiple sources, including basic research, clinical trials, and epidemiological and clinical studies. The available information is frequently fragmented, sometimes contradictory, or simply not available. Thus, a comprehensive assessment of clinical validity and utility requires knowledge synthesis in the form of systematic evidence reviews from multiple sources. Most laboratories are currently not equipped to conduct such analyses.

We believe that a remedy for this problem is a process of systematic knowledge synthesis followed by evidence-based recommendations by an independent group. The Centers for Disease Control and Prevention (CDC) has explored the feasibility and methods for such a process by 2 synergistic initiatives. The first initiative, the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) project was launched in 2004 [2]. The main goal of EGAPP is to establish a rigorous, systematic, evidence-based process for evaluating genetic tests that are in transition from research to clinical and public health practice. An independent, non-federal EGAPP working group developed methods for reviewing evidence on emerging complex genetic tests by modifying existing approaches used by professional organizations, advisory committees, independent task forces (e.g., U.S. Preventive Services Task Force and CDC's Task Force on Community Preventive Services) and international health technology assessment groups. The EGAPP working group also developed processes for identifying, prioritizing and selecting topics, performing evidence reviews, and developing recommendations for practice based on the evidence. As of July 2009, EGAPP has published their methods [3], 5 evidence reports and 4 recommendations, with others still in the pipeline [4].

The second initiative is the Genomic Applications in Practice and Prevention Network (GAPPNet), launched in 2009 [5]. GAPPNet is a stakeholder-driven enterprise

KARGER

Fax +41 61 306 12 34
E-Mail karger@karger.ch
www.karger.com

Accessible online at:
www.karger.com/phg

Muin J. Khoury, MD, PhD
Office of Public Health Genomics, Centers for Disease Control and Prevention
1600 Clifton Road
Atlanta, GA 30333 (USA)
Tel. +1 404 498 0001, Fax +1 404 498 0140, E-Mail muk1@cdc.gov

with representation from academia, government, health care, public health, industry, and the community. The network aims to promote systematic review of research findings, support for translation research, and diffusion of high quality information on genomic applications in practice and prevention. One of GAPPNet's goals is the development of an online knowledge base on the clinical validity and utility of genomic applications [6]. When systematic reviews have been conducted by EGAPP or other health technology assessment groups, the knowledge base will provide links to these reviews. For selected genomic applications lacking comprehensive reviews, brief systematic summaries of available evidence will be provided. When no information is available, this will also be noted, which will stimulate research to fill these knowledge gaps. The GAPPNet knowledge base is expected to provide an initial point of reference for what we know and what we don't know, as well as a source for updated information as it is compiled by GAPPNet members from published and unpublished applied research.

Clearly, establishing a genetic test registry is a worthwhile goal; however, the blueprint by Javitt et al. describes a structure without the necessary utilities: that is, the connections to a growing grid of translational research and evidence-based knowledge syntheses that will provide the essential multifaceted data on validity and utility. Creating a registry with credible information requires more than mandating submission of data by laboratories. The methods and products of EGAPP, together with the goals and upcoming activities of GAPPNet, could easily be integrated into the implementation of the genetic test registry to enhance the utility of the registry to providers, consumers, researchers, and policy makers.

References

1 Javitt G, Katsanis S, Scott J, Hudson K: Developing a blueprint for a genetic test registry. Publ Health Genomics 2009; Published online June 29 DOI: 10.1159/000226593.

2 Evaluation of Genomic Applications in Practice and Prevention (EGAPP). Available at http://www.egappreviews.org/, accessed 07/06/09.

3 Teutsch SM, Bradley LA, Palomaki G, Haddow JE, Piper M, Calonge N, Dotson WD, Douglas MP, Berg AO and the EGAPP Working Group: The evaluation of genomic applications in practice and prevention (EGAPP) initiative: methods of the EGAPP working group. Genet Med 2009;11:3–14.

4 EGAPP working group-recommendation statements. Genet Med 2009;11:15–73.

5 The Genomic Applications in Practice and Prevention Network (GAPPNet). Available at http://www.cdc.gov/genomics/GAPPNet/index.htm, accessed 07/06/09.

6 Khoury MJ, Feero WG, Reyes M, Citrin T, Freedman A, Leonard D, Burke W, Coates R, Croyle RT, Edwards K, Kardia S, McBride C, Manolio T, Randhawa G, Rasooly R, St Pierre J, Terry S, and the GAPPNet Planning Group: The genomic applications in practice and prevention network. Genet Med 2009;11:488–494.