Public Health Genomics Highlights 2013

Posted on January 30, 2014 by Muin J Khoury, Director, Office of Public Health Genomics, Centers for Disease Control and Prevention

At the end of each year, we read about top lists of major events, accomplishments, and milestones. These lists are produced by journal editors, institutions and opinion leaders. CDC is no exception. In December 2013, CDC published its top 5 accomplishments for 2013, which include the *Tips* from former smokers campaign, outbreak investigations featuring pathogen sequencing and advanced molecular detection, the Million Hearts® initiative, eliminating healthcare-associated infections, and celebrating 10 years of the global program to fight HIV/AIDS. Similar lists in other areas of science and technology have been published (for example, see Science and Nature.

2013 was a special year in genomics because it marked the 60th anniversary of the discovery of the structure of DNA and the 10th anniversary of the completion of the Human Genome Project. Our top 5 list focuses on events and issues that illustrate how the use of genomic information is making a real impact on improving health and preventing disease in populations. Here are our top 5 highlights in public health genomics for 2013. We are interested in our readers' opinions and input on these issues and others that we may have left out.

1- 50 Years of Newborn Screening: Saving Lives and Preventing Disease & Disability

2013 year marked 50 years of saving lives through newborn screening. Newborn screening remains the largest public health genetics program in the world and is run by public health agencies in all 50 states in the US. Newborn screening identifies more than 30 conditions that can affect a child's long-term health or survival. Early detection, diagnosis, and intervention in more than 12,000 babies every year helps prevent death or disability. Each year, millions of babies in the U.S. are routinely screened for certain genetic, endocrine, and metabolic disorders using a few drops of blood from the newborn's heel, or a point of care test at the bedside.

2- 10 Years of Family Health History: As Important as Ever

2013 marked the 10th anniversary of Family Health History Day on Thanksgiving. Despite the amazing progress in the past decade, family history remains the simplest and most readily available genomic tool for disease prevention and health care across the lifespan. Family members share genes, behaviors, lifestyles, and environments that together may influence their health and their risk of disease. Most people have a family health history of some diseases (e.g., cancer, coronary heart disease, birth defects, and diabetes) and health conditions (e.g., high blood pressure and hypercholesterolemia). Family health history can inform evidence-based preventive services, such as screening for elevated cholesterol and osteoporosis. The updated Surgeon General's *My Family Health Portrait* tool provides consumers with a free and easy way to record their family health information and is published in several languages. Consumers may use this tool to organize their family history information and share it with their family and health care professionals.

3- Pathogen Genomics and Public Health: The Time is Now

Pathogen genomics is already on CDC's top 5 list for 2013, and it is noteworthy on the public health genomics list. While the prospects for using human genome-based testing in clinical care and prevention are exciting, the emergence of powerful sequencing and bioinformatics tools has completely changed the landscape in the public health fight against infectious diseases. There are numerous applications for pathogen genomics including diagnosing infection, investigating outbreaks, describing transmission patterns, monitoring antimicrobial resistance, and developing interventions such as vaccines. The new field of metagenomics promises to uncover entire communities of microorganisms, often including species never before cultured in the laboratory, that may be detected and characterized, opening the door to understanding the role of environmental, animal, and human microbiomes in health and disease. In 2013 CDC launched the Advanced Molecular Detection (AMD) Initiative, which aims to build critical molecular sequencing and bioinformatics capacities at national and state levels to support public health efforts to control infectious diseases.

4- Whole Genome Sequencing in Healthcare: Great Promise and Great Challenges

We are now formally in the era of Next Generation Sequencing (NGS) which includes many applications such as exome sequencing, gene panels, and whole genome sequencing (WGS). This advanced technology is increasingly utilized to identify genetic causes of rare, uncharacterized diseases, particularly childhood conditions. In addition, tumor-based genomic sequencing is beginning to permeate oncology with major advances in molecularly- targeted tumor classification and gene-directed therapy. In 2013, the Blue Cross Blue Shield Technology Evaluation Center evaluated the clinical use of exome sequencing in the diagnosis of rare diseases. The report showed a significant uptake of exome sequencing into clinical laboratory practice. However, despite the promise of sequencing, there are significant challenges to its general implementation that require additional research and development. These include analytic validation, workforce challenges, and ethical issues related to reporting genomic findings.

5- Policy and Legislative Actions Affecting Implementation of Genomic Medicine

In 2013, several branches of the US government enhanced implementation of validated genomic applications through significant policy and legislative activities while curtailing the premature use of such technologies. These included: the Supreme Court ruling on gene patents; the FDA authorization of the first, next-generation DNA sequencer; the FDA's increased regulatory activity over the direct-to-consumer genetic testing industry; the Affordable Care Act provisions for coverage of some genetic counseling and testing; and the progress in implementation of genomics objectives in the Healthy People 2020 initiative. These actions signal an increasingly robust policy and evidentiary framework for the appropriate use of genomics to improve health.

Finally, to facilitate integration of evidence-based genomics into healthcare and public health, CDC released an ongoing list of genomic applications based on levels of evidence and has promoted a proactive approach for the implementation of "tier 1" applications through partnerships and programs in public health departments. A clickable state map of public health activities was released in 2013 and an implementation tool kit will be released in early 2014 to help public health departments implement selected tier 1 genomic applications that could reduce morbidity and mortality in about 2 million people in the US. An increasingly active public health involvement in genomic implementation will help reduce health disparities, increase appropriate use of technologies, and potentially reduce healthcare costs.

This is our top 5 list for public health genomics highlights in 2013. We have only begun harnessing the power of genomics to improve health and prevent disease and we look forward to progress in the field for many years to come. We invite our readers to submit their input and feedback on this list and other topics in public health genomics.

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