Genomics and Population Health Action Collaborative

The Genomics and Population Health Action Collaborative (GPHAC) is an ad hoc activity under the auspices of the Roundtable on Genomics and Precision Health at the National Academies of Sciences, Engineering, and Medicine. In late 2015, GPHAC recruited a diverse group of stakeholders with an interest in integrating genomics at the population health level. The goal of the group is to explore the opportunities for genetics and genomics research and applications to improve public health, reduce health disparities, and promote genomic literacy. Now in its second year the action collaborative includes three parallel working groups:

- **Population Screening** — led by Jim Evans, University of North Carolina at Chapel Hill and Michael Murray, Geisinger Health System;
- **Cascade Screening** — led by Heather Hampel, the Ohio State University and Katherine Wilemon, the FH Foundation;
- **Implementation** — led by Debra Duquette, of the Michigan Department of Health and Human Services and David Chambers, National Cancer Institute.

The three working groups are developing an online resource guide (“toolkit”) that will serve as a framework for action by public health systems at the state level and health care organizations that are planning to integrate genomics and precision medicine approaches into their current programs.

**Activities to Date:** In the first year, Action Collaborative participants focused on evidence-based genomics approaches that offer an opportunity to improve the early detection and clinical care of individuals with pathogenic variants that cause Hereditary Breast and Ovarian Cancer (HBOC) and Lynch syndrome (LS). Using a cancer-focused case study, an Evidence Working Group examined the current methods used to identify and assess the clinical actionability of evidence-based genetic tests for HBOC and LS. The group then developed a series of written summaries that review the process of horizon scanning and evidence assessment by various consortia including ClinGen, the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) initiative, and the United States Preventive Services Task Force (USPSTF). The Evidence Working Group went on to explore the potential health impact of implementing genomic applications in public health programs by examining modeling strategies, population data, and existing evidence. In April 2017, the group published its final product on the action collaborative website.

The Implementation Working Group is performing information gathering interviews with representatives of state health departments to better understand the barriers and facilitators to public health-based genomics programs that focus on hereditary cancer. The group has also developed a set of outcome metrics that can be used to measure the efficacy of cancer genomics programs at the state public health level. Lastly, a subset of the Implementation group is preparing manuscripts on health disparities related to hereditary cancer syndromes, and outlining possible approaches for public health genomics programs to alleviate those disparities.

**Looking Ahead:** As GPHAC enters its second year, the group will continue to add resources to the online toolkit. The group will expand its focus from state public health systems to include health care delivery systems. Participants will begin to explore genomics-based approaches for familial hypercholesterolemia (FH), another Tier 1 application, in addition to HBOC and LS. Two new working groups will begin including one that focuses on cascade screening, the process by which screening takes place in first degree, at-risk relatives of patients who have a particular condition and another that explores collaborative research and protocols on the benefits and challenges associated with population-based opportunistic genetic screening programs such as the MyCode study run by Geisinger Health System and the GeneScreen study in North Carolina.