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 third full year, the action collaborative has had four parallel working groups:

- **Cascade Screening** — Heather Hampel, the Ohio State University and Katherine Wilemon, the FH Foundation, co-leads
- **Evidence (until 2017)** — Ned Calonge, President and CEO, Colorado Trust, lead
- **Implementation** — Debra Duquette, of the Michigan Department of Health and Human Services and David Chambers, National Cancer Institute, co-leads
- **Population Screening** — Jim Evans, University of North Carolina at Chapel Hill and Michael Murray, Yale University, co-leads

Together, these working groups are developing resources that will serve as a framework for state-level public health systems and health care organizations that are planning to integrate genomics and precision medicine approaches into their current programs.

**Activities to Date:** In its 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, the 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 describe 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 paper on the action collaborative website1.

The Implementation Working Group performed information gathering interviews with representatives of approximately a dozen state health departments to better understand the barriers and facilitators to public health-based genomics programs that focus on hereditary cancer. The discussions from those interviews are in the process of being written up for a paper. The group has also developed a set of outcome metrics2 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.

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In 2017, the Action Collaborative expanded its focus from state public health systems to also include health care delivery systems. Participants also began exploring genomics-based approaches for detecting familial hypercholesterolemia (FH), another Tier 1 application, in addition to HBOC and LS. Two new working groups formed—one focused on cascade screening (the process by which screening takes place in first degree, at-risk relatives of patients who have a particular condition) and the other on population-based genetic screening programs.

Participants in the Cascade Screening working group published a literature review in early 2018 on methods for delivering cascade screening for hereditary conditions. Members of this group are also examining patient preferences and HIPAA policies related to direct contact of at-risk relatives. Lastly, a small team within this group is developing an implementation framework for cascade screening programs and has plan to publish this work.

The Population Screening working group participants developed a paper titled A Guide for Considering Genomics-Based Screening Programs in Healthy Adults, which will be published later in 2018. The paper covers several topics including: optimal genes, settings for screening, informed consent, engagement strategies, ethical considerations, and economics/sustainability.

**Working Group Leads:**

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

**Action Collaborative Participants (past and present):**

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- Hana Bangash, Mayo Clinic
- Seth Baum, Foundation for Preventive and Integrative Medicine
- Erica Bednar, MD Anderson Cancer Center
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- Deborah Bowen, University of Washington
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- Toby Citrin, University of Michigan School of Public Health
- Mindy Clyne, National Cancer Institute
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- Debra Lochner Doyle, Washington State Department of Health
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