

New Strategies in Public Health Genomics



Actions to Save Lives Now

Conference Report



**New Strategies in Public Health Genomics:
Actions to Save Lives Now**

September 7, 2012

CDC Roybal Campus

Atlanta, Georgia

Conference Report

Edited by Lan Q. Le, MPH and Jessica Skiba, BS

Funding for this project was made possible by the Centers for Disease Control and Prevention through McKing Consulting Corporation, contract 200-2009-F-32675. The contents are solely the responsibility of the authors and do not necessarily represent the views of CDC.



December 5, 2012

We are pleased to share this report of the meeting, *New Strategies in Public Health Genomics: Actions to Save Lives Now*. The meeting was held on September 7, 2012, and included 80 participants representing all major sectors of health care and public health involved in providing and advocating for genetics/genomics services for common chronic diseases.

Conference speakers provided background information on three conditions (Hereditary Breast and Ovarian Cancer; Lynch Syndrome; and Familial Hypercholesterolemia) and the evidentiary basis for their use to prevent early disease and death. Break-out groups developed recommendations on how public health programs can be initiated and strengthened in partnership with other sectors of the health system. Representatives of support groups shared recommendations from the perspective of patients and consumers who have experienced these diseases.

The CDC Office of Public Health Genomics would like to thank the Center for Public Health and Community Genomics at the University of Michigan School of Public Health and the Genetic Alliance for their help in planning the event and preparing this report. In the next weeks to months, our office will be posting on our website a toolkit containing useful information and links to resources to assist public health agencies and their partners in planning and implementing these essential programs.

We will be pleased to respond to comments, inquiries and suggestions relating to this report and the public health programs that it recommends. All inquiries should be sent to genetics@cdc.gov

Sincerely,

Muin J. Khoury, MD, PhD
Director, Office of Public Health Genomics

Table of Contents

<i>Content</i>	<i>Page</i>
Opening Remarks - Ursula Bauer	1
Meeting Purposes and Goals - Muin Khoury	2
Introduction to Tier 1 Programs	3
Lynch Syndrome Presentation Summaries	
Speaker 1: Heather Hampel	4
Speaker 2: Debra Duquette	5
Post-Presentation Q&A	6
Hereditary Breast and Ovarian Cancer Presentation Summaries	
Speaker 1: Mark Robson	7
Speaker 2: Amber Roche	9
Post-Presentation Q&A	11
Familial Hypercholesterolemia Presentation Summaries	
Speaker 1: James Underberg	12
Speaker 2: Joan Ware	13
Post-Presentation Q&A	14
Patient and Community Perspectives Panel Discussion	15
Break-out Sessions Reports	20
Familial Hypercholesterolemia Break-out Group	21
Hereditary Breast and Ovarian Cancer Syndrome	
Break-out Group 1	32
Break-out Group 2	39
Lynch Syndrome	
Break-out Group 1	48
Break-out Group 2	55
Afternoon Plenary and Concluding Remarks	64
Appendices	
Agenda	66
Conference Planners	67
List of Participants	68
Speakers' Biographies	71
Panelists' Biographies	74

Opening Remarks

Ursula Bauer, PhD, MPH, Director of the National Center for Chronic Disease Prevention & Health Promotion (NCCDPHP), welcomed participants to the meeting and to the CDC. Chronic diseases are responsible for seven of every 10 deaths in the United States and afflict approximately 140 million Americans. Such conditions cause major limitations for nearly one in 10 of our citizens and account for 75% of the 2 ½ trillion dollars that this country spends every year on medical costs and care. Tobacco use, poor nutrition and physical inactivity are three major risk factors which can be modified to prevent chronic disease. The evidence-based prevention strategies supported by the NCCDPHP are cost effective and even cost-saving, prevent needless suffering and save lives.

In the realm of public health genomics, knowing your family history and use of appropriate genetic testing can also reduce morbidity and mortality from chronic diseases such as *BRCA1/2* associated hereditary breast/ovarian cancer, Lynch syndrome and familial hypercholesterolemia. Those working in public health genomics can learn from other chronic disease programs about ways to change the context and make healthy choices easy for Americans – to reach the greatest number of people at risk and have the largest health impact.

Dr. Bauer reminded us of the four areas of focus through which the NCCDPHP achieves its goals – epidemiology and surveillance; environmental approaches; health systems and community clinical linkages – and challenged the group to consider all four arenas during their deliberations today. She will look forward to hearing the recommendations that emanate from this meeting so that genomics can become more thoroughly integrated into the Chronic Disease program's activities and so that we can better collaborate on improving and saving lives impacted by chronic diseases.

Meeting Purposes and Goals

Muin Khoury, MD, PhD, Director of the Office of Public Health Genomics (OPHG) set the stage for the meeting. He informed the group about a recent conference on “Priorities for Public Health Genomics, 2012 – 2017” and the need to focus on evidence-based interventions that can reduce morbidity and mortality due to chronic diseases. Using the CDC’s “winnable battles” concept as a model, the OPHG has classified public health genomics applications into Tiers I, II and III to help those working in states and localities to begin working on evidence-based interventions that can save lives now. The purpose of today’s meeting is to learn from each other’s efforts to date, foster collaborative partnerships, and develop specific recommendations that will optimize implementation of public health programs in hereditary breast / ovarian cancer syndrome, Lynch syndrome and familial hypercholesterolemia. Similar strategies are expected to be modifiable for use with other diseases that reach Tier I status in the years ahead.

Dr. Khoury’s PowerPoint presentation can be found here:

<http://genomicsforum.org/index.php?note2=khoury>

Introduction to Tier 1 Programs

Three pairs of speakers provided an introduction to the public health programs that can address hereditary breast and ovarian cancer syndrome (HBOC), Lynch syndrome (LS) and familial hypercholesterolemia (FH). For each pair, the first speaker was an expert on the disease/condition as well as the utility of the genetic test associated with the condition: Heather Hampel, The Ohio State University, presented on LS; Mark Robson, Memorial Sloan-Kettering Cancer Center, presented on HBOC; and James Underberg, NYU School of Medicine and NYU Center for Prevention of Cardiovascular Disease, presented on FH.

The second speaker was a public health professional with experience implementing the screening program utilizing the testing described by the first speaker. Debra Duquette, Michigan Department of Community Health, presented on LS; Amber Roche, Public Health Seattle & King County, presented on HBOC; and Joan Ware, National Association of Chronic Disease Directors, presented on FH. More information about the speakers can be found in the Appendix.

All speakers were advised to gear their 15 minute presentations to the likely knowledge of public health professionals, providing information that would be useful to these professionals in developing and implementing the programs being discussed.

A 5 minute Q&A session followed each pair of presentations.

Presentation Summaries

LS Speaker 1

Heather Hampel, MS, CGC
The Ohio State University

LS is the most common heritable cause of colorectal cancer (CRC) and endometrial cancer (EC). One out of every 35 CRC patients and one out of every 40 EC patients has LS.^{1,2,3} CRC cancer risk varies from 15% for individuals with *MSH6* or *PMS2* mutations to 56% for those with *MLH1* & *MSH2* mutations. In contrast, the general population has a CRC risk of 5%. Additionally, within the general population, the average age of diagnosis of CRC is approximately 70 but for individuals with LS, the average age of diagnosis is approximately 45.

LS can be screened through microsatellite instability (MSI) testing or immunochemistry staining (IHC) of tumor tissue. If a presymptomatic individual has been identified with LS and undergoes colonoscopies starting at the recommended age and at the recommended frequency, CRC incidence and related mortality can be reduced. An added benefit of identifying LS in CRC patients is the prevention of a second CRC. Individuals with CRC have a 16-30% chance of developing a second primary CRC during the 10 years after their first diagnosis. For a person with LS, the surveillance recommendation is more aggressive than in those without LS. The National Comprehensive Cancer Network (NCCN) guidelines recommend that individuals with CRC and LS have a colonoscopy every 1-2 years for life while those without LS should be screened 1 year after diagnosis, repeat in 2-3 years, and then every 3-5 years based on findings.

Cascade testing for relatives of CRC patients with LS can lead to the identification of unaffected carriers and positively impact public health outcomes. A longitudinal study of asymptomatic family members of individuals with LS conducted by Jarvinen et al (1995 and 2000) in Finland found that colonoscopy screening at 3 year intervals reduced the overall mortality in family members of LS patients by 65%.^{4,5} In the United States, assuming three affected relatives per proband, cascade testing has the potential to identify an additional 12,345 individuals at high risk who can consider earlier and more intensive screening.

Lives can be saved by diagnosing LS early. Universal screening for LS among newly diagnosed CRC patients is feasible and cost-effective. Limiting LS screening to individuals who meet the Bethesda Guidelines or Amsterdam II Criteria would miss 25% of CRC patients and 65% of EC patients with LS. This year, 16,460 Americans can be identified with LS through universal

¹ Hampel et al. (2005). Screening for Lynch syndrome (hereditary nonpolyposis colorectal cancer). *New Engl J Med*, 352 (18), 1851-1860.

² Hampel et al. (2008). Feasibility of screening for Lynch syndrome among patients with colorectal cancer. *J Oncol*, 26 (35), 5783-5788.

³ Hampel et al. (2006). Screening for Lynch syndrome (hereditary nonpolyposis colorectal cancer) among endometrial cancer patients. *Cancer Res*, 66, 7810-7817.

⁴ Jarvinen et al. (1995). Screening reduces colorectal cancer rates in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology*, 108 (5), 1405-1411.

⁵ Jarvinen et al. (2000). Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology*, 118 (5), 829-834.

screening of newly diagnosed CRC patients and through cascade testing of their relatives. Ms. Hampel's PowerPoint presentation can be found here:

<http://genomicsforum.org/index.php?note2=hampel>

Lynch Syndrome Speaker 2

Debra Duquette, MS, CGC
Michigan Department of Community Health

Today, approximately 12 out of 400 individuals being diagnosed with CRC will have LS. It remains unclear how many of those individuals and their relatives are being screened and diagnosed. The hope for universal LS screening of individuals with newly diagnosed CRC is to enhance cancer prevention and screening for patients and their families, detect and prevent cancer earlier, reduce health care costs, and save lives.

The 2009 Evaluation of Genomic Applications in Practice and Prevention (EGAPP) recommendations for genetic testing for LS state that there is sufficient evidence to recommend offering genetic testing for LS to individuals newly diagnosed with CRC in order to reduce morbidity and mortality in relatives.⁶ Healthy People 2020 embraced the EGAPP recommendations, and increasing the proportion of newly diagnosed CRC patients who receive genetic testing for LS is listed as one of the genomics objectives. However, national data are needed in order to measure whether the objective will be met. One way to obtain national data on LS is to collect MSI test results through state cancer registries. An optional tumor specific element was introduced in 2010 and the CDC's Division of Cancer Prevention and Control is currently piloting a study in select states looking at the collection of that data element by cancer registries.

Since 2008 the state of Michigan has been funded by the CDC to conduct surveillance on LS. Michigan conducted telephone surveys, reviewed Medicaid claims data, performed medical chart audits, and provided screening for low income individuals in three counties with high CRC mortality rates. Through these efforts it was discovered that the majority of individuals at risk for LS did not know that a genetic test was available. Additionally, LS screening was performed in less than 2% of CRC cases according to an audit of 2006-2010 medical records. Furthermore, no health plan in Michigan has a policy which aligns with the EGAPP recommendations; most policies are aligned with the NCCN recommendation, the Bethesda Guidelines, or the Amsterdam II Criteria.

While there are many barriers to implementation of universal LS screening of individuals newly diagnosed with CRC, LS is a genetic condition that brings clinical, public health, and advocacy groups together. In future, the implementation of LS screening may be used as a model of how to build local, regional, national, and international collaboration. But starting today, there are steps that state health departments can take to address LS. Among them are increasing public and provider awareness and knowledge about the condition, integrating LS screening into CRC screening programs, and utilizing existing state data. Efforts to implement and expand universal

⁶ Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group. (2009). Recommendations from the EGAPP Working Group: Genetic testing strategies in newly diagnosed individuals with colorectal cancer aimed at reducing morbidity and mortality from Lynch syndrome in relatives. *Genet Med*, 11, 35–41

screening programs are currently under way through the Lynch Syndrome Screening Network (LSSN). LSSN promotes universal LS screening and facilitates the implementation of LS screening across different institutions through the sharing of data and protocols.

Ms. Duquette's PowerPoint presentation can be found here:

<http://genomicsforum.org/index.php?note2=duquette>

Post-Presentation Q&A

The questions posed to the speakers during the 5 minute session centered on the lessons that can be learned from the Columbus (Ohio) Hereditary Non-polyposis Colorectal Cancer (HNPCC) study, providers' and the public's awareness of LS, and whether metrics and model legislation language for LS exist.

In Ms. Hampel's experience with LS testing and cascade screening, many families may be interested in testing once they are informed about their risk, but it is quite possible that a vast majority of families do not know that a risk exists. Additionally, in a research setting it is often easy to reach relatives because counseling and screening can be performed in the home. However, when implementing cascade testing on a large scale via the clinic, it may be difficult to have people come to a centralized place to receive counseling and testing. Patients may be deterred by logistical issues such as long driving distances and inconvenient parking.

In terms of policies, there is currently no model legislation for health reform involving LS in any state and no state has even attempted to pass any legislation about this issue. One participant noted that it is important that model language for LS legislation be prepared and readily available so that it can be inserted into large legislative proposals.

Metrics need to be developed which can be used to demonstrate the cost-effectiveness of the testing, the number of lives likely to be saved, and the impact on quality of life. While quality of life has not been factored into most such calculations, a cost-effectiveness analysis conducted by Mvundura et al. (2010)⁷ showed that LS screening is cost-effective in terms of incremental costs per life years saved.

One participant reported that not many health maintenance organizations (HMOs) are collecting cost efficiency data because 85%-95% of the reimbursement for testing is through the hospital diagnostic related groups (DRGs) reimbursement. Hospitals are compensated for the episode of care at a set amount of money regardless of the testing they perform. In most cases hospitals are losing money related to doing extra LS testing under the DRG. Hospitals with a high percentage of Medicare patients stand to lose even more money.

Finally, in regards to increasing provider awareness about LS testing and cascade testing of family members, the American Medical Association, in conjunction with the National Coalition for Health Professional Education in Genetics (NCHPEG), has created a web-based, for-credit, continuing medical education module.

⁷ Mvundura M, Grosse SD, Hampel H, et al. (2010). The cost-effectiveness of genetic testing strategies for Lynch syndrome among newly diagnosed patients with colorectal cancer. *Genet Med*, 12(2), 93-104.

HBOC Speaker 1

Mark Robson, MD
Memorial Sloan-Kettering Cancer Center

In 2005, the US Preventive Services Task Force (USPSTF) recommended that women with a family history associated with *BRCA1/2* mutations be referred for genetic counseling and evaluation for *BRCA* testing.⁸ Kurian et al demonstrated that the prevalence of *BRCA1/2* mutations in women with breast cancer is non-trivial.⁹ Zhang et al found that in invasive ovarian cancer, nearly 13% of women have a mutation in *BRCA1/2*.¹⁰ While there are other genes associated with breast cancer, *BRCA1/2* mutations are the most common.

Family history is a major risk factor for breast cancer. Pioneers like Henry Lynch and Mary-Claire King were the first to link breast and ovarian cancer together as well as to identify a hereditary predisposition to HBOC. Those with a family history are at a significantly increased relative risk for developing breast cancer and the degree to which their risk increases is dependent on the age their relative was diagnosed and the number of affected relatives. This increased risk can be attributed to factors such as random aggregation, i.e. having a large family with many older women, shared socio-cultural risk such as age of first childbirth, and shared environmental factors. However, incomplete penetrance makes recognition of hereditary disposition challenging. The detection of a *BRCA1/2* mutation can have a substantial impact on relatives. For example, unaffected carriers who are between 40-50 years old have a 1-2% per year risk of developing breast cancer and a lesser but still substantial risk of developing ovarian cancer.

Unlike LS, there are no intermediate tests like MSI or IHC which can identify *BRCA*-associated malignancies, as *BRCA1/2* associated cancers do not have clearly distinguishable characteristics. The identification of a *BRCA* mutation has a significant impact on affected women because of the management choices that will be presented to them. The NCCN has clinical criteria for *BRCA* testing. However, it is difficult to translate recommendations into algorithms.

The management of HBOC ranges from preventative surgeries such as prophylactic mastectomy to surveillance options such as breast MRI and mammograms. The diagnostic yield of MRI is higher than mammography and the combination of MRI and mammography is superior to mammography alone. In comparison to breast cancer, ovarian cancer tends to present at an advanced stage when treatment is likely to be less successful and the mortality rate is very high. Transvaginal ultrasound is used to detect ovarian cancer but the detection often occurs when the cancer is advanced. The recommendation for women with *BRCA1/2* mutation is risk-reducing salpingo-oophorectomy once childbearing is complete. Statistical models

⁸ US Preventive Services Task Force. (2005). Genetic risk assessment and *BRCA* mutation testing for breast and ovarian cancer susceptibility. *Ann Intern Med*, 143,355-361.

⁹ Kurian et al. (2009). Performance of Prediction Models for *BRCA* Mutation Carriage in Three Racial/Ethnic Groups: Findings from the Northern California Breast Cancer Family Registry. *Cancer Epidemiol Biomarkers Prev*, 18, 1084-1091.

¹⁰ Zhang et al.(2011). Frequencies of *BRCA1* and *BRCA2* mutations among 1,342 unselected patients with invasive ovarian cancer. *Gynecol Oncol*, 121(2), 353-7.

can be used to estimate the impact of the various HBOC interventions. Allison Kurian, MD, MSc and Sylvia Plevritis, PhD of Stanford University have developed a decision tool for women with a *BRCA* mutation.¹¹ Using a Monte Carlo model to calculate the impact of different interventions for women with a *BRCA* mutation, Kurian et al demonstrated how various management choices can impact an affected woman's probability of survival.¹²

Dr. Robson's PowerPoint presentation can be found here:

<http://genomicsforum.org/index.php?note2=robson>

¹¹ <http://brcatool.stanford.edu/>

¹² Kurian et al. (2010) Survival analysis of cancer risk reduction strategies for BRCA1/2 mutation carriers. *J Clin Oncol*, 28(2), 222-231.

HBOC Speaker 2

Amber Roche, MPH **Public Health-Seattle and King County**

The Healthy People 2020 Genomics objective has set a 10% improvement in the proportion of women with a family history of HBOC who receive genetic counseling as a goal. Family history can be integrated into current public health breast cancer screening and detection methods in order to achieve this objective. The state of Washington's Breast, Cervical, and Colon Health Program (BCCHP) is funded by the CDC, the State of Washington, and the Susan G. Komen Foundation. The program helps low-income uninsured and underinsured clients obtain breast, cervical, and CRC screenings.

There are two options for integrating family history collection into current breast cancer screening programs. Either a focused or a broad approach can be taken to identifying women with *BRCA1/2* mutations and their relatives. Under the former, public health departments can start with women who have been diagnosed with a *BRCA1/2* mutation, identify whether there is a family history, then make referrals to genetic counseling, and finally recommend cascade screening for their relatives. The second approach involves reviewing the records of all clients in a state's BCCHP program and also asking providers to identify patients within their practice who are at risk of developing HBOC. Electronic medical records (EMRs) may be a useful tool for collecting family history. The state of Washington has also made efforts to integrate family history collection into breast cancer screening by revising the BCCHP history and exam form to include family history questions such as the number of male relatives with breast cancer, the age at which a relative was diagnosed, etc. and promoting the use of the Cancer Family History Guide and an online tool, the Breast Cancer Genetics Referral Screening Tool (B-RST), developed by Cecelia Bellcross, PhD, MS, CGC.¹³

Based on Public Health-Seattle and King County's BCCHP's experience, there are a few challenges to integrating family history into breast cancer screening programs. These include the alignment of data collection forms with data systems and operational challenges such as having an adequate amount of time and number of personnel to review and run reports related to client and family history.

Additional challenges to HBOC screening and testing include ensuring that providers and clinic staff are knowledgeable about *BRCA1/2* testing and genetic counseling referral. Education is important because inappropriate referrals and testing have been cited by genetic counselors as an issue. There is also the challenge of ensuring coverage and access to genetic counseling for underinsured and uninsured clients once a *BRCA1/2* mutation has been identified. As changes to the Affordable Care Act are underway, it is important for public health departments to verify if genetic counseling and testing will be included in their state's essential health benefits.

Washington's BCCHP operates under a decentralized model wherein the funding first goes through the state department of health and is then distributed to prime contractors such as

¹³ <http://www.brcagenscreen.org>

Public Health-Seattle and King County. If HBOC screening is implemented on a statewide level, this regional model may add an extra layer of complexity to implementation plans as each region has its own agencies with their own rules. However, a regional model allows local public health departments to focus on the needs of their own population.

Ms. Roche's PowerPoint presentation can be found here:

<http://genomicsforum.org/index.php?note2=roche>

Post-Presentation Q&A

The discussion following the presentations by Dr. Robson and Ms. Roche centered on HBOC genetic testing referrals and insurance coverage. One of the challenges mentioned in Ms. Roche's presentation was inappropriate ordering of genetic tests. The topic sparked discussion about the need for physician education, recognition of genetic counselors as providers by CMS, and what is deemed appropriate vs inappropriate testing.

It was reported that data from Myriad Genetics regarding HBOC testing in the state of Washington showed that 30% of referrals for HBOC testing came through genetics clinics, and 70% came from physician offices. However, these data do not demonstrate a correlation between appropriateness and the source of the referrals. It was remarked that "appropriate" genetic testing and ordering can be a loaded term which requires definition; it is important to not make the assumption that appropriate orders are more likely to come from one source over another. What can be gleaned from the data is that the majority of orders are not coming from genetics specialists. Since the majority of orders do come from outside of genetics clinics, there is a need to provide education to physicians about HBOC testing. A related issue is the role of genetic counselors. One participant commented on the need for CMS to recognize genetic counselors as reimbursable entities.

A major barrier for patients is the issue of insurance coverage and the cost of tests. Medicare does provide coverage for HBOC and LS testing; however, HBOC genetic testing coverage only extends to women who previously had breast or ovarian cancer, or in the case of LS, the patient must have a personal history of uterine or colon cancer. Due to this gap in coverage, an unaffected woman who is for instance at a 50% risk of developing HBOC, is of Medicare age, and is eligible for Medicare will not be covered for genetic testing. One participant remarked that the genetics community needs to make sure that legislators close the existing loopholes and protect the rights of patients. The upcoming change in health care legislation is an opportunity to require protection for the patient and serves as a reminder of the need to extend coverage for high-risk individuals.

As a point of clarification, it was noted that to qualify for breast screening through the state of Washington's BCCHP, women ages 35-39 must have breast symptoms. Women with a family history of HBOC and no clinical symptoms may not qualify based on their family history alone.

FH Speaker 1**James Underberg, MD, MS****NYU School of Medicine and NYU Center for Prevention of Cardiovascular Disease**

FH is an inheritable, autosomal dominant disorder associated with an increased risk of cardiovascular disease (CVD). FH is usually caused by a mutation in the low-density lipoprotein receptor gene (*LDLR*) but also can be due to a mutation in the *Apo B* and *PCSK9* genes. Clinical signs of FH include severe hypercholesterolemia, xanthelasma, corneal arcus, and tendon xanthomata. A history of premature coronary heart disease and family history of CVD are also good indicators of FH risk.

Heterozygous FH is not a rare genetic condition and is as common as Type 1 diabetes. The worldwide prevalence of heterozygous FH is approximately 1 in 500 and in the US, there are approximately 620,000 individuals with FH.¹⁴ The prevalence of FH is higher in founder groups such as South African Afrikaaners, French Canadians, Christian Lebanese, and Ashkenazi Jews.¹⁵ Within these groups, the prevalence of FH can be eight times greater than in the general population.¹⁶

Despite the prevalence of FH and the available treatment options, the disease is underdiagnosed. The three main criteria currently being used to diagnose FH are 1) the Simon Broome Register in the United Kingdom (UK); 2) Make Early Diagnosis Prevent Early Death (MEDPED) in the US; and 3) the Dutch Lipid Clinic Network (DLCN) in the Netherlands. Screening recommendations and the use of genetic testing in screening for FH also vary from country to country. In the US, the National Lipid Association (NLA) recommends screening all individuals by age 20 and using one of the three sets of diagnostic criteria to identify index cases. The next step is to locate and offer cascade screening to relatives.

Genetic screening for FH has important implications for disease management for probands and their relatives. An FH patient will have a 20 times greater chance of having a CVD event in their lifetime compared to someone in the general population. Once identified, FH patients can start on statin therapies which have been shown to significantly reduce an FH patient's risk for myocardial infarction.¹⁷ Furthermore, a study in the Netherlands has demonstrated that cascade screening for family members of probands can lead to an increase in usage of cholesterol-lowering treatment.¹⁸ Identifying a parent with FH also has implications for their children. While intervening during childhood may prevent CVD events in adulthood, pharmacological treatment of FH in childhood is a controversial topic among parents and medical professionals. There are concerns about the long-term effects of medication, the lack of outcomes data, and misdiagnosis. Dr. Underberg's PowerPoint presentation can be found here:

<http://genomicsforum.org/index.php?note2=underberg>

¹⁴ Goldberg et al. (2011). Future issues, public policy, and public awareness of Familial Hypercholesterolemias: Recommendations from the National Lipid Association Expert Panel on Familial Hypercholesterolemia. *J Clin Lipidol*, 5, 133-140.

¹⁵ Naoumova, R. (2004). Current management of severe homozygous hypercholesterolaemias. *Curr Opin Lipidol*, 15 (4), 413-422.

¹⁶ Austin, MA. (2004). Genetic causes of monogenic heterozygous familial hypercholesterolemia: a HuGE prevalence review. *Am J Epidemiol*, 160, 407-420.

¹⁷ Verschmissen et al. (2008). Efficacy of statins in familial hypercholesterolaemia: a long term cohort study. *BMJ*, 337, a2423.

¹⁸ Umans-Eckenhouse et al. (2001). Review of first 5 years of screening for familial hypercholesterolaemia in the Netherlands. *Lancet*. 357, 165-168

FH Speaker 2**Joan Ware, BSN, MPH
National Association of Chronic Disease Directors**

Since FH is an underdiagnosed condition, family health history collection can be a powerful method for identifying index cases and implementing cascade screening among their relatives. An example of a cost-effective program that has used a family history tool is the Family High Risk Program (FHRP). From 1983-1999, the Utah Department of Health, local health departments, the University of Utah and Baylor College medical schools, the Utah State Board of Education, and local school districts implemented a program for identifying families at risk of heart disease, diabetes, cancer, and stroke. High school students enrolled in 10th grade health education classes were given a Health Family Tool (HFT) and asked to collect up to three generations of family health history. The health histories of families that consented to participate in the FHRP were then analyzed for disease risk. High-risk families were contacted and offered family based interventions.¹⁹ An evaluation of FHRP demonstrated that health behavior changes occurred in both high-risk and average-risk families.

Today, the use of an electronic version of the HFT can be promoted among providers and patients. Awareness of the effectiveness of family health history collection among providers is needed, especially in the case of FH. In terms of the public, family health history collection can be promoted at events such as the American Heart Association's Heart Walk.

Unlike LS and HBOC, there is no Healthy People 2020 objective for FH. However, public health can convene stakeholders and educate both providers and the public about FH in order to identify probands and their relatives. The creation of an FH registry and the use of EMRs are needed in order to increase surveillance efforts. Collaboration and partnerships across stakeholder groups are needed in order to identify the approximately 600,000 Americans with FH who have not been identified.

Ms. Ware's PowerPoint presentation can be found here:
<http://genomicsforum.org/index.php?note2=ware>

¹⁹ Johnson J. et al. (2005) Utah's Family High Risk Program: bridging the gap between genomics and public health. *Prev Chronic Dis* [serial online] 2005 Apr. Available from: URL: http://www.cdc.gov/pcd/issues/2005/apr/04_0132.htm

Post Presentation Q&A

The FH Q&A discussion centered on questions about identifying high-risk individuals at an earlier age and the safety of statin use in children.

The first question posed to Dr. Underberg and Joan Ware was about the age-related prevalence of FH. Dr. Underberg said that the cholesterol cut-off levels vary based on age. The concern is that instead of age being the risk factor for FH, age is the multiplier of risk. When examining patients' cholesterol levels, it is better to treat patients early, rather than waiting for them to become higher risk. This is problematic because physicians are used to using risk-based algorithms in determining whom to treat for CVD. In other countries, it is typical for FH patients to be considered high-risk from Day 1. Hopefully, in new iterations of US guidelines, FH will be considered a coronary artery disease risk equivalent. It was also noted that there is no ICD-9 code specifically for FH, which complicates the process of identification and diagnosis of FH by a physician.

The second question posed to the speakers was about whether there is evidence demonstrating the safety of long-term statin treatment for children. To Dr. Underberg's knowledge, there are data from European studies showing delays or regressions in carotid intermedial thickness in children taking statins. However, pharmacological treatment for children is a controversial topic. In the management of FH for children, it is still essential to keep in mind the need to use interventions which encourage healthy lifestyle choices such as smoking cessation in parents of affected children and reduction of other high-risk behaviors.

Patient and Community Perspectives Panel

The morning plenary session concluded with a panel discussion moderated by James O’Leary from Genetic Alliance. The discussion focused on the challenges and opportunities associated with screening implementation from the perspective of stakeholders at the community, patient, and consumer level. The 45 minute presentation centered on issues related to clinical intervention, policy, education, and surveillance.

Panelists: Sue Friedman, FORCE: Facing Our Risk of Cancer Empowered; Sabrina Ford, Michigan State University; Winona Hollins Hauge, Governor’s Interagency Council on Health Disparity (Washington State); National Community Council Genomics SPIG/University of Washington Health Promotion Research Center; Cristi Radford, Lynch Syndrome International; Rochelle Shoretz, Sharsheret: Your Jewish Community Facing Breast Cancer; Katherine Wilemon, FH Foundation

Policy: Development of partnerships

How can the actions of state public health agencies be better coordinated with those of community and patient advocates? What barriers exist that could prevent these groups from working together effectively?

Reaching out to the community before an intervention is developed in order to build synergy and effective deliverables

- Departments of health where genomics is a main focus should reach out to patient advocacy and community groups during the program development stage rather than wait until the implementation stage. Inviting the community and patients to the discussion table when the program is still on the ground floor will help develop goodwill, collaboration, and mutual respect. Rochelle Shoretz said, “Even more important than getting the word out to the communities is having the word of the communities at the very, very beginning of the strategy and policy discussions.”
- Academic researchers may also benefit from taking the approach above because researchers often bring in community partners based on the needs of an RFA rather than developing a proposal with the input of the community partner.
- States can use a “three amigos” approach wherein leaders representing the state, patient/community, and medical or legal fields meet and strategize about how to best develop and implement culturally appropriate health programs. The leaders’ different perspectives ensure that the voices of multiple stakeholders are being considered during the program development phase.

Going beyond the public health comfort zone

- Public health practitioners should be amenable to a paradigm and strategy shift. Winona Hollins Hauge suggested approaching community partners with a message of, “We would like you to come and help us reach out to your community and we have no preconceived notions about what this is going to look like.”

Sharing of expertise

- In addition to utilizing the knowledge of community partners to develop focused and effective programs for target populations, local, state, and federal governments can share their expertise and experiences with non-profit organizations. The contributions of time, advice, and networking can be a valuable incentive for patient and community groups to work with state, local, and federal agencies. An example of this collaboration is the CDC's Breast Cancer in Young Women initiative which has provided funding to organizations such as Sharsheret and Sisters Network Inc. to develop evidence based interventions which can be rolled out as model programs helping young women understand and navigate cancer screening, diagnosis, and treatment.

Policy: Identify fiscal & personnel resources

Unlike newborn screening, the testing that we've discussed today requires insurance coverage and reimbursement in most states. How big of a barrier is reimbursement for testing in your communities? In what instances does that coverage contradict public health recommendations?

Mismatch between insurance coverage guidelines and LS and HBOC recommendations

LS

- Insurance coverage for germline mutation analysis for LS varies depending on whether the payer's guidelines stipulate that the patient be 50 years of age or under and meets the Amsterdam II Criteria or the Bethesda Guidelines. In her experience working with hospitals to set up LS screening, Cristi Radford has had success in helping hospitals obtain reimbursement for IHC testing but has found that obtaining coverage for germline mutation analysis and follow-up tests such as *BRAF* mutation analysis or hypermethylation testing remains an issue.

HBOC

- Not all state Medicaid programs offer coverage for *BRCA1/2* mutation testing.
- Medicare coverage for *BRCA1/2* mutation testing is limited to women who have had a prior history of breast cancer in addition to meeting other stipulated criteria. Under these restrictions, unaffected relatives interested in HBOC testing would not be covered.
- The NCCN's Breast Screening and Diagnosis guidelines recommend that women with increased risk of breast cancer begin screening at the age of 25. However, the screening may not be covered by some payers.

Resources for the underinsured or uninsured

- The FH Foundation is working with private vendors to decrease the out-of-pocket cost of lipid panels for patients.
- The Cancer Resource Foundation, Inc. has recently launched a national cancer genetic testing copay assistance program for patients who cannot afford their copay or deductible.
- Patient Services, Inc. offers MRI screening for young women who are at high risk for breast cancer.
- Myriad offers a hardship program for *BRCA1/2* testing to uninsured patients.

Clinical Intervention: Assuring competent public and health care workforce

How big are the gaps in healthcare and public health from your perspective, both in terms of training in genetics and genetics providers? Are the trends positive or negative?

Awareness and education

- The panelists agreed that there has been an increase in public awareness of the three conditions over the past decade. However, as visibility for LS and HBOC screening tests becomes more prominent in the public eye, both physicians and patients must be educated about the availability, specificity, sensitivity, and validity of screening tests such as IHC and *BRCA1/2* mutation testing in order to prevent negative outcomes such as incorrect ordering of tests, misinterpretation of test results by physicians, false assurance about risk, or the election of unnecessary preventative surgeries by patients.
- Unlike the genetic testing used for HBOC and LS, an FH diagnosis can be made using an inexpensive lipid panel. However, underdiagnosis of the condition remains a central issue. Katherine Wilemon said, “There is a need for education, even within the medical community, and within the public conversation. Patients themselves are unaware of what a dire risk it [FH] is.”
- Awareness about the conditions among healthcare workers may help young patients feel more comfortable and less frustrated when they undergo their first testing or screening procedure.
- One way to decrease the gap between healthcare and public health is to increase awareness about the utilization of genetic experts such as geneticists and genetic counselors. Sue Friedman, said, “A lot of people know that if they have heart disease they should see a cardiologist or if they have an eye problem they can see an ophthalmologist but they don’t understand that there are genetic experts out there that are available to them.”

Clinical Intervention: Evaluating effectiveness, accessibility, and quality

In working with your communities, how big of a barrier is fear to getting people to utilize services? Are there any issues specific to hereditary conditions or genetic testing?

Mistrust of genetic testing

- Sue Friedman referred to an online survey of FORCE and patients from The Ohio State University, wherein the top two reasons patients gave for not undergoing genetic testing were cost and fear of discrimination. Survey takers also reported a lack of awareness of the Genetic Information Nondiscrimination Act (GINA) and expressed concern about discrimination by insurance companies. Survey takers indicated that if they had questions about GINA, they would likely ask their physician. However, not all physicians are knowledgeable about GINA.
- Mistrust surrounding genetic tests can also stem from the type of dialogue patients and providers have about genetic testing. Providers should be educated about how to broach the topic in a way that will not exacerbate the fear and the myths surrounding genetic testing.

Education: Informing and educating consumers

What barriers (including cultural, stigmatizing, family structure, religious practice, etc.) complicate the process of reaching family members? What strategies should be employed where those barriers are high? What resources are available to surmount them?

Barriers

- *Over assurance.* Some families may feel secure in the education they have received about condition specific prevention and intervention options. Depending on the health literacy of the family, Sabrina Ford said that education may sometimes provide an “artificial sense of security” because patients may feel confident in their knowledge level about a condition and thereby not examine their family history.
- *Family history.* Public health practitioners and physicians need to recognize the diverse types of family structures when developing cascade screening programs or collecting family history. For some, a family history is not easily traceable. Therefore, understanding how family structures differ can really help providers shape the conversations about cascade testing in a more effective way.
- *Family dynamics.* Long standing power struggles between siblings or other relatives may lead to resistance to cascade testing. For example, what one relative may perceive as encouragement may be construed as pressure by another.
- *Cultural dynamics.* The ease of conversations about cancer and disease can take different forms depending on the community. Reaching some communities may also be a challenge because of mistrust. For example, Sabrina Ford cited difficulties with recruiting Latina participants for the *Kin Keeper* program because of the fear of deportation.
- *Cultural concerns.* Reluctance to discuss family history may be related to concern about a relative’s reputation. For example, in some Jewish communities there exists a fear that a genetic abnormality can be deemed a black mark on the family which will negatively affect the marriageability of the children. Additionally, for some African Americans, the Tuskegee Study continues to perpetuate mistrust.
- *Religious concerns.* Discussing family history may seem irrelevant to some patients because they may believe that God’s will is a more powerful determinant of disease risk than family history.

Strategies

- Education should include disease specific information about how to have a dialogue with family members about cascade testing and family history. James O’Leary noted that it is important to examine what types of education are being provided to patients and their families. He said, “Education isn’t just about the gene or the condition, it is about how you have a conversation with a family member about health issues.”
- A public service announcement or campaign may ease families into a conversation about screening and testing.
- An important component of the public health strategy is ensuring that there is buy-in from the target communities and not making the assumption that every community is in fact the same.

The panel discussion ended with the panelists expressing excitement about heading into the break-out sessions to discuss the toolkit and action plan amongst a diverse group of stakeholders.

Break-Out Sessions Reports

Conference participants were divided into five pre-assigned break-out groups. Each group included those with expertise specific to the disease/condition, together with members of each sector that needed to be involved in developing effective programs (i.e., public health agency; medical care; health systems; academe; advocacy and/or support groups; disease-specific organizations; community). The charge for each group was to identify:

1. Strategies to achieve buy-in and the initial decision to develop and implement an action plan.
2. Elements of an effective action plan for implementing the screening program.
3. Fiscal, personnel and educational resources needed for effective programs.
4. The components of a “toolkit” that would be most useful to public health professionals developing these programs.
5. Sectors that need to be included in the program and how best to elicit their participation.
6. Anticipated challenges and how to address them.

Note: Breakout group summaries are a compilation of individual opinions and may not necessarily represent general agreement among group members.

Break-Out Session: Familial Hypercholesterolemia

Facilitator: Summer L. Cox, Oregon Health Authority

Notetaker: Jessica Skiba, University of Michigan School of Public Health

Participants:

Donna Arnett, American Heart Association

Jean Chabut, Michigan Department of Community Health

W. Gregory Feero, Dartmouth Medical School

Rebecca Giles, Utah Department of Health Asthma Program

Alan Gilstrap, Genzyme

Scott Grosse, CDC

Yuling Hong, CDC

Paul Hopkins, MEDPED

Stephan Kopecky, Mayo Clinic

Michael Shapiro, Oregon Health & Science University

James Underberg, NYU School of Medicine and NYU Center for Prevention of Cardiovascular Disease

Joan Ware, National Association of Chronic Disease Directors

Katherine Wilemon, FH Foundation

Selvi Williams, Kaiser Permanente Center for Health Research

I. Initial Buy-in

A. Action Plan Goal

The action plan needs to address the issue that even though people are screened for cholesterol, the diagnosis of FH is often not made or is overlooked. By not properly diagnosing FH patients, nothing can be done in terms of screening or treatment of family members. In order to address this issue, an action plan focusing on reduction of morbidity and mortality is necessary. The main target for the use and implementation of the action plan includes, but is not limited to, public health practitioners, with the idea of creating a plan to implement at public health levels (national, state, and possibly local) screening for FH through utilization of partnerships and resources.

B. Strategies

- Model FH after other chronic diseases that have gained attention in recent times.
 - Utilize public health as the convening power.
 - Use public health professionals to lead and organize the effort.
 - Tie FH into American Heart Association activities & promotions, or those of other organizations/efforts
- Identify a “champion” who can help make FH visible, important, and relevant.
 - Use personal stories
 - Patient stories are likely to be the most powerful tool because FH is a silent disease but can cause significant morbidity and mortality.
 - Make the case for FH more compelling through use of data and the recognition of cost-saving and lifesaving interventions.
- Include children
 - Clarify that the main reasons for population-wide cholesterol screening in children is to find and treat FH because there are interventions available that can be lifesaving.
 - Advocates and communities like programs that help children, which may be a way to secure buy-in if information about pediatric FH is presented.
 - NHANES has produced some data on children and cholesterol levels, which should be presented to bolster community support.
- Use what we have
 - Existing and current resources should be utilized and geared toward FH screening and identification of FH patients.
 - There are preliminary data that may assist in identifying patients; they may guide additional research necessary for buy-in.
 - Needs assessments and further research may be necessary because it is unknown what percent of Americans have had their cholesterol levels tested, regardless of age. The CDC should create a clickable map showing percentage of individuals having cholesterol screening by state. *[Editorial*

note: such a map actually exists; see:

http://www.cdc.gov/dhdsp/data_statistics/fact_sheets/fs_cholesterol.htm]

- In order to gain support for implementation of FH screening, more data need to be collected and reported to show the benefit to the individual first identified with FH, not just the benefits that can be achieved through cascade screening.
- Patients can be rallied to demand change from political representatives, employers, payers, and physicians for better care and reduction of morbidity and mortality. Policy can arm advocates in order to make strides in FH screening.
- Build more support
 - Because FH screening is similar to other translations of genetic research into action, other plans and strategies may be applicable to secure buy-in. For instance, there are commonalities shared among FH, LS AND HBOC which may be necessary to use in terms of buy-in and policy making.

II. Action Plan Elements

A. Policy

Issues and challenges

- Screening for FH and subsequent treatments for FH are expensive and need to be paid for by insurers.
- Quality assurance measures for FH are needed to ensure that quality of care is the best possible.
- There are issues integrating screening measures into clinical practice, and FH screenings need to be somehow incorporated into already existing screening protocols.
- There is not a unified voice or unified front for quality of care for FH, so nothing is able to get done for improving diagnosis and treatment. The number of diagnoses needs to be increased because screening increases treatment, and treatment improves health outcomes.
- Malpractice suits occur for failure to diagnose FH.
- The role of genetics and genetic testing needs to be decided.

Strategies

- Cost coverage
 - Insurers need to pay for screening and testing.
 - A diagnosis of FH should automatically allow for coverage of appropriate medications and prescriptions, as indicated by the NICE guidelines.
 - The coverage would need to include potent statins and medications for LDL regulators.
 - Existing policies for renal failure may serve as a guide for making similar policy for FH.
 - Physicians should be reimbursed for cascade screening.

- Cascade screening should be publicly funded. The Netherlands has a model program that we could consider.
- Set metrics and quality measures
 - There need to be policies to ensure that practices are maintaining the highest quality through quality assurance measures.
 - Accountable care organizations need to be employed. National Committee for Quality Assurance (NCQA) reporting metrics should set policy metrics. National Quality Forum (NQF) is also a key organization for quality assurance.
 - Highlight centers of excellence and reward centers that meet suggested guidelines for FH patients to improve the standard of care. The Cystic Fibrosis Foundation has a model program that could be modified for this purpose.
- Incorporate cholesterol screening into existing screening.
 - Link cholesterol screening to other required health services, such as immunizations in children prior to entering schooling.
 - It could be possible to systematically incorporate cholesterol testing at other major points when 9-11 year olds are seen by health care providers such as:
 - Gardasil and other immunizations
 - Athletic exams
 - Use state mandated triggers to send educational materials to patients at specific times. For instance, a positive pregnancy test triggers providers to inform patients about folic acid for the pregnant woman and pertussis vaccinations for the male partner.
- Use a well-known person who is affected by FH to champion for cascade screening coverage.
- Create coalitions made up of different sectors and public health organizations.
- Malpractice suits can be limited through creating appropriate policies and screening criteria to be followed by healthcare providers.
- The future of genetics in policy and public health still needs to be decided.

B. Clinical implementation

Issues and Challenges

- There are not optimal referral pathways and a large proportion of patients self-refer.
- There are several different guidelines and recommendations, which all differ slightly in terms of screening criteria.
- The requirements for screening children need to be clarified.

Strategies

- Create optimal referral pathways.
- It may be practical to consider a change in health care models so that a person who is not a physician is responsible for systematic review of family history of all early-onset disease.
- There should be a unified screening protocol and diagnostic criteria.
 - The USPSTF and EGAPP have existing recommendations for other conditions such as HBOC and LS, so it is necessary for them to create recommendations

for FH that are unified, given that there are several different recommendations currently available.

- Indicate to the USPSTF and EGAPP the need to create recommendations for 1) specific clinical scenarios for screening targeted by age (especially pediatric); 2) specific screening protocol and 3) diagnostic criteria.
- It is important to encourage the USPSTF endorsement of screening cholesterol levels in appropriate patients.
- The uniform diagnostic criteria need to be easily accessible and freely available on the internet.
- The single set of criteria may be based on lipid levels or family history.

C. Surveillance

Issues and Challenges

- Currently there are no patient registries or programs which use EMRs to flag possible patients.
- Data are not shared and are typically held within universities and institutions.
- EMR-based programs cannot diagnose families and EMR-based trigger programs cost roughly \$1.2 million for statin prescriptions.
- Patients are virtually invisible because we do not know where they are, so we cannot best serve them.
- HIPAA and privacy issues may emerge when registries are used for data collection or case finding.
- There are limited data available currently.

Strategies

- Utilize state level health information exchange (HIE) to detect potential FH patients, establish a registry system, and monitor health outcomes.
 - Through use of the registry, there should be state mandated lab reporting of cholesterol over a certain level or threshold through flagging to physicians.
 - An educational message about FH with information about available resources should be sent to both physician and patient.
- Creation of a national registry. This can become a reality through working with the Office of the National Coordinator for Health (ONC) and health system administrators.
 - The registries should utilize basic surveillance data techniques from the CDC passed down to the states.
 - There should be one single FH registry which can be used to find index cases and subsequent cases through cascade testing.
 - The registry should be a hybrid of sorts, with separate patient and physician portals and separate patient and physician entry.
 - Data can be shared using a registry and can allow for the required additional research and better learning. The registry can be validated for research.
- Genotyping in different areas of the country is required to better understand need.
- Utilize EMRs

- Because EMR-based programs cannot diagnose families, there must be a flagging mechanism put in place in order to find cases. This mechanism should send secondary information about FH to physicians to ensure that a diagnosis is made.
 - Currently, there are existing programs for other conditions, such as diabetes. EMRs need to have meaningful use standards to indicate or suggest FH.
- Identify patients with premature coronary heart disease events through EMRs and death certificates or other records in order to evaluate them against FH criteria. In addition, when an index case is identified, implement cascade screening of family members.
- Certain measures will need to be implemented in order to ensure that HIPAA is not violated and patient privacy is upheld.
- Existing data need to be analyzed and made available.

D. Education

Issues and Challenges

- People need to know and understand FH because there is a lack of familiarity with and understanding of the condition. Physicians need to educate patients.
- Health care providers and medical professionals involved in recognition and diagnosis of FH cases are not always informed and often don't know what to look for in the screening process.
 - Physicians are unaware of FH diagnosis and the genetic epidemiology of FH.
 - Family history is often overlooked.
- There are many barriers to overcome in terms of working with and educating patients, including education levels and health literacy, familial and cultural norms, bias, and language barriers.
 - Patients need to understand their results.
 - Patient support is needed.
 - Patients are able to refer themselves if they have the knowledge.
 - It is often difficult to send information intended for patients because it is ignored or not understood/made relevant.
- Research institutions should be included in the education process.

Strategies

- Patient Education
 - Patients need to understand what their results mean for them.
 - Information related to FH patient care needs to be explained to patients.
 - The probability of developing FH can be explained to patients using diagrams with proportions instead of percentages.
 - Health care providers and industry could work together to use education mechanisms that bring patients together.
 - Using patients as educators encourages patient empowerment.
- Patient support groups may need to involve more than just the health care team, consider including faith-based support and community leaders.

- Public awareness campaigns and media campaigns should be created to educate the general population.
 - Research is needed to identify which groups need what kind of education and what the best way is to get the right messages out to the public.
 - The CDC and states can and should develop materials and fact sheets for patient education, in addition to distributing existing educational materials.
 - The general messages need to be uniform.
- Educate health care providers
 - Educate health care providers and physicians regarding the genetic epidemiology of FH, including the value of family history as an assessment tool.
 - Clinical awareness and training programs need to be developed, including educational materials on how to identify new and potential cases and how to proceed after a potential case is identified.
 - Education regarding the genetic epidemiology of FH needs to reach general practitioners, pediatricians, dermatologists, OB/GYNs, and other key health care employees in order to ensure that all cases of FH are diagnosed. Physicians often assume that all patients with FH have all the cardinal symptoms or they may look for cases similar to those reported in textbooks, which tend to focus on the most significantly affected individuals
 - Physicians need to be alerted to the symptoms and warning signs they have missed.
- Mechanism to educate both providers and patients
 - Educate physicians and patients on a broader scale about how to communicate with family members. Help them learn how to talk with families and how to teach families to talk to each other. There are many different dynamics within families, which pose a challenge in conversations about an FH diagnosis.
 - Physicians and laboratories should send treatment and education materials to the patient because then plans are implemented and followed through.
- Patients can self-refer and should be empowered to take control of their own health and life to the extent possible.
 - Use patients as the starting point for diagnosis; consider patient-oriented web-based diagnostic tools like the registry from MEDPED.
- Academic institutions need to present findings in meaningful and significant ways in order for the general public and physicians to understand findings.

III. Toolkit Recommendations

A. Buy-in

- *Coalitions.* The formation of coalitions and partnerships is critical to implementing screening and increasing diagnosis of FH. An integrated approach led by experienced public health professionals who are able to direct and organize a joint effort will be highly successful.
 - There is a need to “make the case” for why public health needs to be part of FH stakeholder groups and use their convening power.
 - Funding & involvement from the federal government, written into larger cooperative agreements that include FH work, is especially needed in the

formation of partnerships and task forces in order to make progress in FH screening. Partnerships and coalitions need to include:

- Patients
- Health care providers, including but not limited to primary care physicians, cardiologists, nurses (RN, LPN, BSN) and nursing organizations, obstetricians and gynecologists (OB/GYN), registered dietitians (RD), pediatricians, dermatologists, Physician Assistants (PAs), musculoskeletal specialists, lipidologists, phlebotomists, medical pathologists, and receptionists
- Non-profit and patient and patient advocacy organizations, such as The FH Foundation, Genetic Alliance, International FH Foundation
- Professional and medical societies and organizations, such as the American Heart Association (AHA), American Medical Association (AMA), American Academy of Pediatrics (AAP), NHLBI, American College of Clinical Chemists, Association of Public Health Laboratories, NLA, American Board of Clinical Lipidology
- Health systems administrators
- Private payers and Medicaid. Medicaid in Michigan just decided to cover lipid screening for children at age 2 and then as early teenagers.
- Industry, such as companies that produce screening tests
- Research and academic institutions.

B. Policy

- *Single criterion.* A single unified criterion to be the most predictive of health risks should be endorsed, such as MEDPED or Simon-Broome.
 - The USPSTF and EGAPP have existing recommendations for other conditions such as HBOC and LS, so it is necessary for them to create recommendations for FH, including a clarified focus on screening children.
 - The current NHLBI guidelines for sickle cell anemia may serve as a guide to creating guidelines for FH.
 - The NICE guidelines should also be consulted.
 - Family history assessment tools should be developed as part of the new criterion.
- *Patient stories.* Patient stories are vital to secure buy-in. The necessary community support can be drawn in through personal connections, and then backed up with data. Patient stories should include stories about family history, personal history, and pediatric cases.
 - *Heart UK* trains highly motivated patients to present to local members of Parliament. This allows for translation to members of government to respond to community need.
 - If a patient champion (and possible legislator) is identified, it may be possible for legislation to be enacted on behalf of FH. For example, Senator Tim Johnson (D-SD) had a preexisting condition, Hereditary Hemorrhagic Telangiectasia (HHT), and nearly died. He realized the federal government was not doing anything for HHT, so he sponsored the Hemorrhagic Telangiectasia Diagnosis and Treatment Act of 2011.

- Personal testimonies like those of FH Foundation founder, Katherine Wilemon, who learned about her high cholesterol at age 15, had a heart attack at 39, and was subsequently diagnosed with FH can be very applicable.

C. Clinical Intervention

- *Best Practice.* It is important to consider best practices from other countries, including updated evaluations of the health impact and cost of the Dutch and United Kingdom's FH screening programs, when formulating US policies. Create cascade screening projects based on existing international programs in Brazil, Canada, and the Netherlands. These projects are publicly funded screening programs, which may be necessary for FH.
- *Pedigree Analysis.* Pedigree analysis is cost effective. Tools should be developed to alert health care providers about family history of FH. These tools can play a major role in cascade testing.
- *Universal screening.* It may be beneficial to have universal screening in some age group to capture affected individuals. European and three American professional groups (NLA, NHLBI, and American Academy of Pediatrics) recommend universal screening for children ages 9-11.
 - It is cost-effective because testing all prior to onset of disease lowers the burden of disease.
 - There needs to be a way to capture a population and schools are an ideal place to begin.
 - The most high risk children that can be detected at an early age can exhibit drastic health improvements.
 - Because parents play a major role in maintaining their child's health, they also can influence healthy behaviors at a younger age. Children at this age are more amenable to change.
 - The USPSTF recommends screening for cholesterol by age 20.
 - Tie in cholesterol screening at other major points in children's lives at ages 9-11 years
 - Gardasil, other vaccines are administered
 - Lead testing
 - Athletic exams
 - Physical examination for entrance to school
 - Tie in cholesterol screening with other major events in the lives of adults
 - Recommend screening of male partners after female partner's positive pregnancy test. Cholesterol screenings of pregnant women may find higher than normal levels of cholesterol due to pregnancy and thus is not advised.
 - Follow-up screening at age 20

D. Surveillance

- *Registries.* Create national registries.

- *Existing research.* Report existing supporting data about FH, including current literature reviews and current publications and make documents readily available. Factsheets should be created for patient education.
- *EMR.* EMRs should have programs to flag potential patients and charts should be screened more thoroughly.
- *Children.* Use school systems as an entry point to start because a majority of children attend schools; this provides a great way to capture potential cases and extend into cascade screening. Health classes in high schools should teach about family history and use “trees” as a tool for collecting family health history data.

E. Education

- *Utilize workforce.* Empower employees such as receptionists or phlebotomists to flag charts and possibly identify FH patients. This can be done through additional training and seminars, and could possibly be incorporated into physician education as well.
- *Educate patients.* Ensure that educational information is accurate, current, distributed, and available to all patients and potential patients. Self-referral is important for diagnosis.
 - Inform current and potential FH patients about the availability of resources. Resources may be available and produced by industry and patient organizations, such as:
 - Genzyme’s FH Journeys & FH Journeys on YouTube:
<http://www.fhjourneys.com/>
 - Learn Your Lipids: Learnyourlipids.com
 - Materials generated by CDC
 - Materials generated by states
 - Factsheets
 - Literature Reviews targeted to patients
- *Community education.* Coalitions and public health should partner with state chapters of the AHA to promote health education at community events such as Go Red for Women and Heart Walks.
- Community awareness and public health messages should be delivered as unified messages.
- *Physician Education*
 - Incorporate FH into Grand Rounds to enhance medical student and resident education.
 - Utilize popular medical journals, such as the Journal of the American Medical Association (JAMA) or the New England Journal of Medicine (NEJM).
 - Reviews of current literature and fact sheets are needed for physician education.

Break-out Session: Hereditary Breast and Ovarian Cancer – Group 1

Facilitator: Winona Hollins Hauge, Governor's Interagency Council on Health Disparities,
National Community Committee Genomics SPIG/ University of Washington Health Promotion
Research Center

Notetaker: Elizabeth Schmitt, Emory University graduate student

Participants:

Sylvia Au, Hawaii State Department of Health,

Cecelia Bellcross, Emory University School of Medicine

Sara Copeland, HRSA

Lori Farmer, International Society of Nurses in Genetics

Sue Friedman, FORCE: Facing Our Risk of Cancer Empowered

Sandra Fryhofer, AMA Council on Science and Public Health

Jane Korn, Minnesota Department of Health

Kimberly Lewis, Georgia Center for Oncology Research and Education

Dana Meaney-Delman, Emory University School of Medicine

Mark Robson, Memorial Sloan-Kettering Cancer Center

Debbie Saslow, American Cancer Society

Katrina Trivers, CDC

Cynthia Vinson, National Cancer Institute

I. Initial Buy-In

The group discussed how to get buy-in from the appropriate public health and health system decision-makers. Community engagement can influence these decisions, and advocacy organizations, some of which are already working toward these goals, can elicit community engagement.

A challenge which can lead to significant limitations in obtaining buy-in at all needed levels is a lack of funding. Public understanding, acknowledgement, and support will fuel collaboration and help recruit the right people and advocacy organizations that not only can assist in achieving these goals, but also will provide financial and organizational support that will help enable larger scale initiatives toward change.

In addition to public organizations, hospitals also need to buy-in on an organizational level. Health care is changing -- educating administrators as well as physicians is important. In order for the action plan to be successful it is not only the health care system that needs to buy-in; health care administration must also be involved in order to achieve the desired changes.

Through public support and collaboration, using pre-existing resources and tools, HBOC screening on a public health scale can become a reality. In hopes of seeing such coverage occur on a national level, all state, regional and local public health agencies need to agree with these goals, and this buy-in can begin by encouraging collaboration between outside experts and relevant public health staff. Once HBOC screening has achieved public health support, insurance coverage will have to follow, making HBOC risk evaluation and screening a standard of care opportunity provided to at-risk individuals and families.

A. Action Plan Goals

The break-out session setting was “an opportune time to address patients at high risk for breast and ovarian cancer based on personal and family history.” Defining the goals of an action plan is an essential step toward an efficacious session. In doing so, several questions arise: What do we *really* want: High specificity or sensitivity? What should be the intervention public health works toward: counseling or testing? Is there anything wrong with telling some people that they do not need testing? Ultimately, in hope of avoiding “inappropriate” or unnecessary testing, a public health concern referred to in the morning presentation by Amber Roche, the goal was defined as follows:

The goal of the action plan is to increase the awareness of genetic counseling as a resource, properly referring more at-risk patients, and ultimately improving the availability of such testing across the population. Sensitivity rather than specificity should be considered when selecting criteria for referral of patients and families for such services.

In the event that this goal is achieved, counseling and accurate risk assessments would increase, enabling public health to guide a larger number of appropriate patients towards

genetic testing, and testing would be provided to unaffected family members through cascade screening.

B. Strategies

- With the goal of better evaluating the risk of patients who have personal and family histories through increasing the appropriate utilization of genetic counseling, an important strategy is improving the current rhetoric on what constitutes genetic counseling. Specifically, the language surrounding genetic counseling needs to be adjusted, better defining *what* genetic counseling is and *who* is qualified to do genetic counseling, as well as adding information about the availability of cascade testing to a positive genetic test following appropriate genetic counseling. Adding such language to the USPSTF guidelines on HBOC screening is a very powerful means of announcing nationally the importance of promoting the availability of genetic counseling and HBOC screening.
- We need a process to measure the efficacy of genetic counseling. This includes providing evidence that such counseling is a cost-effective intervention.
- Education was described as the most impactful resource available, and needs to be a key element of the strategic approach taken by a multi-disciplinary team of HBOC screening champions. Taking the steps of thoughtfully educating the patient and consumer community, the health care community, the policy-makers, and ultimately the public health community, will help achieve the desired goal of testing the at-risk population.

II. Action Plan Elements

A. Policy

Issues and Challenges

- Much of the discussion of barriers related to the current lack of recognition of genetic counselors. Health care providers and public health practitioners, administrators and members of the public need to better understand and acknowledge the value of genetic counseling and, further, genetic counselors should be recognized as CMS providers.
- Coverage is needed for counseling, testing, follow-up screening, and other recommended preventative measures. Medicaid and Medicare need to recognize the need for such coverage as well.
- Until there are more comprehensive CPT codes specific to genetics, it is very difficult for health systems to know what they have paid for, due to coding discrepancies and the absence of specific billing codes.
- Issues related to cost-effectiveness will have to be discussed and managed in further policy conversations, and may call for adjustments in EGAPP recommendations as well.
- Patients have a deep mistrust of insurance companies. They fear having genetic information in their medical records due to concerns about being labeled by certain genetic information. This is a very real anxiety in the consumer population and needs to be carefully considered when developing an action plan and formulating policy.

Strategies

- In order to achieve the desired policy changes, there is a need for partnerships.

- The community needs to be engaged in policy-making efforts.
- Genetics staff needs to be a part of state public health programs.
- Champions in insurance companies need to be involved with the push for coverage expansion.
- Obtaining licensure for genetic counselors in all states is an essential step toward such recognition, and coverage is likely to follow if this is achieved.
 - Licensed genetic counselors should have the opportunity to order genetic tests. Without recognition of genetic counselors and the benefits of genetic counseling, it will be difficult to expand coverage.
 - With the intervening element of genetic counseling for risk evaluation, more appropriate referrals for testing will follow.
- Recommendations should be aimed at reducing disparities; men and women from all backgrounds should be offered coverage and appropriate genetic services and testing by trained professionals to maximize efficacy and surveillance of patients at risk for HBOC.

B. Clinical Intervention

Issues and Challenges

- The multi-factorial discrepancies in patient care became a concern that was discussed in depth. The group discussed the real challenges patients face in using the health care system while being subject to racial and socioeconomic related discrepancies.
- Patients also bring their own cultural taboos, mistrust of the medical system, and fears of stigmatization.
- Physician biases in combination with individual patient backgrounds and concerns give rise to many discrepancies that need to be carefully and systematically addressed.

Strategies

- Identifying at risk families, covering their counseling visits and providing cascade testing as needed.
 - Appropriate patients and families can then be referred in greater numbers.
- Research and investigation must continue to reinforce the utility of such clinical practice in order for it to be acknowledged as a standard of care.
 - Increasing evidence and using such data effectively is essential to our ability to apply the action plan appropriately.
- Currently, evidence shows that MRI is a more sensitive screening tool and should be used as a standardized screening method for breast and prostate cancer.
- Increasing awareness of coverage of genetic counseling practices is necessary as more clinical interventions are needed.

C. Surveillance

Issues and Challenges

- There were several ideas and recommendations for surveillance improvement and the major challenge was funding. It becomes very difficult to achieve anything on such a large scale without financial support.

Strategies

- Expand surveillance beyond the responsibility of the government and move into the private sector and corporations.

- Developing an improved means of data sharing and common metrics was considered the most effective strategy to be implemented.
- Improve surveillance through the improvement of the EMR system
 - Develop a standardized tool within the EMR system to flag families with a high-risk family history. This could be a way of improving the tracking of patients who have received genetic counseling and that have a high-risk family history.
 - To help and intervene with these flagged family histories, insurance companies must be involved.
- A family history intake should become a standard part of care, and should be further recorded and tracked via medical records across the population. This can be done through the development of new CPT codes specific to genetics. To have this happen, vendors who make EMRs would need to buy-in.

D. Education

The group unanimously deemed education to be the most important element of the action plan.

Issues and Challenges

- The group discussed the current misconceptions of what risk is – among patients, health care providers, and policy makers alike.
- In the health care system, community education needs to start with primary care.
 - Oncologist Mark Robson commented on his experience with non-oncologists attempting to care for cancer patients and not having the proper reference materials. This leads to discomfort in the physician, and therefore inadequate information and conservative referrals in terms of genetic counseling and testing.
- One of the biggest limiting factors in achieving the desired education goals is the lack of funding, as organizations tend to fund treatment rather than prevention.

Strategies

- Education must be provided to three different audiences: the lay community, health care providers, and policy makers.
 - Utilize personal stories of the many patients who have both suffered from and conquered HBOC. Sharing patients' stories makes a big impact. This strategy can be applied in approaching the development of educational programs for all three target communities.
 - Educators must be knowledgeable about and careful to provide age appropriate information on the importance of HBOC risk.
- Educate the lay community
 - When presenting the lay community with this material, it is important to start with the next generation of men and women entering the recommended age for counseling and screening
 - In hopes of reaching the next generation, it was suggested that such education be integrated into college curricula.
 - Everyone should be informed about the importance of family history and sharing family history. This could be advertised through broadcasting public service announcements and national campaigns, possibly by partnering with other organizations.
- Educate providers

- Working to provide adequate education and increasing primary care providers' competency in evaluation of HBOC risk will enable primary care providers to more effectively refer patients.
- Strategies proposed to effectively educate health care providers involved offering and implementing education modules. Specifically, continuing medical education (CME) and continuing educational unit (CEU) modules for physicians and nurses were recommended as well as "Just in time" education modules for physician use.
- An example of how public health could model new training initiatives and requirements to better educate physicians and other health care providers to refer patients for genetic counseling was the Florida mandatory HIV education requirement for doctors and nurses. Fulfillment of such required education modules in the Florida program was tied to reimbursement and licensure in the state. This model could be a very effective means of ensuring education of health care providers.
- Educate policy makers
 - The education of policy makers would focus on a cost/benefit analysis of implementing HBOC counseling and screening coverage.
 - Hone in on the financial value of making HBOC testing and cascade screening a national public health reality.
- Clarifying misperceptions.
 - There are very common misunderstandings that need to be clarified in educational materials and modules. Men in general are unaware that they are at risk for HBOC.
 - Additionally, the population as a whole is commonly unaware that their paternal family history is just as significant as their maternal family history.
 - In areas where there are not many Ashkenazi Jewish individuals those of that ethnicity may be unaware of their risks or even of their ethnic origin.
 - There is also a dangerous misconception often held by the non-Jewish population that if you are not Jewish, you are not at risk for HBOC.
- Create partnerships
 - Utilize the month of October, currently celebrated nationally as breast cancer awareness month, as a golden time for collaboration with breast cancer foundations as well as public health organizations to educate the lay community. Such partnership provides the possibility of obtaining funding support for educational efforts.
 - Creating partnerships with a variety of consumers, advocates, health care providers, public health representatives, and even policy makers from states that have already approved HBOC coverage, provides not only support and resources, but also allows an educational team to share various perspectives from their different backgrounds and expertise. This ultimately will assist in obtaining policy buy-in, in that the education would not be for and from a single population, but rather for and from several communities, emphasizing the overall importance and recognition of HBOC counseling and screening coverage.
 - A multi-disciplinary team can provide a powerful and diverse work force to approach this plan.

III. Toolkit Recommendations

A. Policy

- The CDC toolkit should include updated guidelines and recommendations for HBOC counseling and testing. These statements should include information relevant to

both male and female at-risk patients, affected and unaffected patients, and should “allow for screening of patients under the age of 40 who do not have access to mammography or MRI.”

B. Clinical Intervention

- There needs to be a targeted tool just for HBOC.
- Develop a flow chart depicting optimal referral pathways. The flow chart could outline the steps of intervention in an understandable way.
- For consumers, the toolkit should also make available family history screening tools for genetic counseling and risk assessment.
 - Such tools should include information for patients explaining what they need to know when collecting and recording their family history. Some family members are reluctant to share family cancer history and sharing such information and recalling such stories can be emotionally difficult as well.
- Include the current and (when available) new USPSTF guidelines in the CDC toolkit. The USPSTF guidelines are evidence-based recommendations and not screening criteria but are very influential. Hopefully, genetic counseling will be included in the recommendations which are being reviewed and edited.
- Emphasis was placed on the need to develop performance measures for health care providers. In order to effectively intervene clinically, there needs to be a means to evaluate physicians so that they are paid for the *quality* of information and care given to patients rather than the *volume* of referrals. Such measures would evolve from a standard of care statement that could also be included in the CDC toolkit.

C. Surveillance

- Tools and registries should be refined and made available on a national level.
- Some existing registries that could be utilized include:
 - Title V Information System
 - Pregnancy Risk Assessment Monitoring System
 - Behavioral Risk Factor Surveillance System (BRFSS)
 - National Program of Cancer Registries
 - Surveillance, Epidemiology and End Results (SEER)
- In addition, the use of previously developed cancer genetics referral screening tools such as the Breast Cancer Genetics Referral Screening Tool should be more widely advertised and made available to the population.

D. Education

- It is important to make appropriate educational resources available to various audiences (lay community, health care community, policy makers and insurance companies).
 - These materials would include thorough explanations of different types of risk, as there are common misunderstandings across all populations.
- Age appropriate risks and risk assessment protocols should be presented in a way that has meaning and is understandable to the layperson, the physician, and the insurance representative in order to improve understanding about the meaning of lifetime and age-specific risk.
- Intercultural Cancer Council: ICCnetwork.org
- National Institute for Health and Clinical Excellence
- Center for Public Health and Community Genomics
- Genetic Alliance
- UW Health Promotion Research Center: Depts.washington.edu/hprc

Break-out Session: Hereditary Breast and Ovarian Cancer - Group 2

Facilitator: Rochelle Shoretz, Sharsheret: Your Jewish Community Facing Breast Cancer

Notetaker: Rachel Webster, Emory University graduate student

Participants:

Beverly Burke, Connecticut Department of Public Health Genomics Office

Wendy Cohn, University of Virginia School of Medicine

Sabrina Ford, Michigan State University

Elizabeth Garner, Myriad Genetics

Karen Greendale, Consultant, McKing Consulting Corporation, contractor for Office of Public Health Genomics, CDC

Katherine Kolor, Office of Public Health Genomics, CDC

Rebecca Nagy, National Society of Genetic Counselors

Patricia Page, Emory University

Amber Roche, Public Health Seattle & King County

Maren Scheuner, VA Greater Los Angeles Healthcare System

Vickie Venne, VA Salt Lake City Health Care System

Barbara Zehnbauer, CDC

I. Initial buy-in

A. Action Plan Goals

In an effort to decide the best strategies to achieve buy-in, this group first acknowledged that there are certain materials necessary to support the need for a HBOC public health approach. These are:

- State Cancer Registry Information
 - States should find out what information is collected by their state cancer registries. Find out whether the registries contain information about family history and/or genetic testing and what steps would be necessary to implement the collection of these data.
 - For states with existing HBOC data, the available information may be used to best estimate the impact of implementing the action plan and to convince state officials of the potential benefits of implementing the action plan.
- Myriad
 - Myriad currently collects data from each of the tests ordered for HBOC. These data include family histories, genetic mutations, demographics, and ancestry. To date, much of the data have not been analyzed. A working relationship with Myriad to analyze such data may be helpful. These collected data may be used to create a report on the benefits of a public health approach, especially in the absence of state cancer registry data.
 - Myriad holds the patent for *BRCA1/2* mutation testing. A public health approach to testing for HBOC would require some type of partnership or a significant partnership with public health to ensure access to testing and evaluation of impact.

B. Strategies

- Supplying funds
 - Providing state health departments with financial incentives may be particularly effective because of the cost associated with implementing the ideas within the action plan. Providing money will give the states the ability to put the action plan in place without having to adjust current budgets.
 - The funding amounts do not necessarily have to be new or large. Several examples were given showing that the items in the action plan may be implemented by partnering with other existing grantees or projects. Additionally, examples were given of projects that were able to accomplish a great deal using money from small grants to cover the initial costs.
- Establishing need
 - For states that have yet to implement any type of public health approach to screening for HBOC, it may be convincing to look at data showing the impact the action plan would have on the state's population. Assessing current and existing state practices and comparing them to the possibilities laid out in the action plan may be very convincing, in addition to using data collected and analyzed from state cancer registries and Myriad.

- Demonstrating cost effectiveness
 - There is a cost benefit to screening women for HBOC. State budget makers may not understand that implementing early screening would save money, especially in the long run. In order to create or incorporate HBOC screening programs into public health it is necessary to use preliminary data to document potential saving and cost-effective measures, even if the savings are conservative.
- Engaging the Public
 - The public must be engaged in efforts to influence state policy makers. There are many HBOC patient support and advocacy groups. These groups, along with any general public support for HBOC, could be used to sway the opinions of elected officials and state health departments.
- Cultural sensitivity
 - Each community is different and a plan that worked in one area may not be as effective in another. States should be sensitive to their target population. By bringing community members to the planning conversation, the individual states will better their programs, as well as improving buy-in at the community level.
- Showing the already prepared toolkit
 - Due to limitations of resources and budgets of state health departments, it is necessary to indicate to states that the toolkit includes tips for implementing an action plan in an accessible and straightforward manner. The toolkit will include a list of resources and examples of how to implement the action plan.
- Release a CDC recommendation
 - The CDC could release a recommendation in terms of HBOC screening. However, this step will likely not be enough on its own, but will add weight to any other argument presented to state public health departments. Adding a nationwide expectation for HBOC screening will give states goals to achieve, and thus more motivation to implement an action plan.

II. Action Plan Elements

A. Policy

Issues and Challenges

- Educating legislators
 - There are large gaps in the education of legislators about HBOC. This deficit is reflected in the laws concerning health care and cancer, which affects population health. Educating these individuals about the services needed by families with HBOC is necessary. For example, educating legislators about patients who are currently not covered by insurance or Medicare may be the first step in improving the insurance laws in each state.
- Financial resources
 - Limited budgets and funding sources produce a shortage of funds available for implementing new programs. In addition, different groups with different agendas are vying for the same limited funding sources.
- Limited Work Force

- There are a limited number of public health officials with genetics training who can carry out the action plan.
- There are also limited numbers of service providers (genetic counselors, genetic nurses, medical geneticists, etc.) to treat patients after they have been identified as being at high-risk for HBOC.
- USPSTF Guidelines
 - The USPSTF guidelines help explain the importance of HBOC on a state level, but they do not include affected women. Hopefully the next version of these guidelines will address this issue.
- Future changes
 - The field of medical genetics is quickly changing and constantly updating the information to be used in health care. Information and recommendations will change in the next few years as research improves, which requires states to stay flexible in terms of changes in policies and procedures.
- State licensure for genetic counselors

Strategies

- Incorporate HBOC language into state cancer plan
 - Many states have some language about HBOC in their state cancer plan, but not all do. Including HBOC language into state cancer plans shows support for and acknowledges the importance of HBOC at the state level. This additional support should make it easier to highlight the importance of the action plan.
- Tie HBOC screening into existing funding or existing projects.
- Use EMR
 - With the push to use EMRs, there should be a family health history tool built into the software used in clinics.
 - Companies should be encouraged to offer a product that will meet meaningful use standards.
 - Clinics should be encouraged to purchase products that will meet their family health history collection needs.
 - Lab and provider representatives should work with vendors to create products that will address the needs of multiple groups. If lab representatives are not consulted, there may be significant mismatch in EMR functionality and user friendliness.

B. Clinical Interventions

Issues and Challenges

- Access
 - The cost of genetic testing can be quite high, especially when insurance companies refuse to cover or pay for screening.
 - Problems with payers may be due to a lack of knowledge about the screening process. Educating insurance companies on the process of genetic testing and its importance could positively affect many patients' access to screening and care as no one screening test is the best fit for every patient and there are multiple commercially available *BRCA1/2* mutation screening tests.

- Genetic counseling could be required for best care and practice, and would avoid screening using inappropriate tests.
- Too many tools
 - Because there are several family health history collection and risk assessment tools available, there is confusion about which ones should really be used.
 - The tools should be evaluated and the identified best practices should be included within the action plan.
 - There was agreement that multiple tools are likely necessary for different applications.

Strategies

- Help providers use time efficient tools
 - State public health departments should be a resource for helping clinicians find the easiest and most time efficient family health history tool for their practice. It may be necessary for state health departments to create a list of appropriate tools.
- Identify existing professionals for referrals
 - Clinicians should have a resource available to help make the proper referrals when a family health history indicates a possible genetic condition.
 - The National Society of Genetic Counselors (NSGC) has a tool for finding genetic counselors within a particular area, and this information should be available on state public health department websites as well.
 - Include a list of local genetic counselors specializing in HBOC.
 - Include a search component for these specialists.
 - Current lists of genetic counselors do not include whether or not the counselor accepts patients on a no fee or sliding scale basis. A list of counselors who accept these patients would be helpful for clinicians referring patients who cannot afford counseling otherwise.
- Quality Indicators
 - The state health departments should be responsible for developing and distributing quality indicators for HBOC recognition in clinics. States should encourage quality indicator use.
 - One step to ensuring quality HBOC care in clinics is to be sure that all clinicians are aware and educated on the USPSTF recommendations regarding HBOC. Without knowing these recommendations, it would be difficult for providers to adequately identify and refer at-risk patients.
- Encourage use of Surgeon General's Family History Tool
 - Clinicians should encourage the use of this tool at home as well as in their offices.
 - Setting up a station in the waiting room could give patients the opportunity to present the provider with a detailed family health history before the appointment, instead of hoping the patients remember to bring the forms to the next visit.
 - In offices without resources to support the in-office family health history collection, simply asking patients to use the tool at home may be the best route.

- Including educational material on the tool could make the patient more apt to use it.

C. Surveillance

Issues and Challenges

- State cancer registries
 - Using state cancer registries to monitor HBOC screening and diagnosis is ideal. Unfortunately, for many states, there is a lack of HBOC screening and diagnosis data available or within state registries.
- Provider participation
 - It is necessary to involve providers in surveillance efforts. Using the current tools and protocols, providers should be willing and able to enter appropriate patient data for state surveillance use.
 - Providers also need to be better educated on taking an accurate family history if they feel unqualified to do so. This education should include all providers, not just OB/GYNs and oncologists.
 - While encouraging health care providers to participate, quality indicators should also be monitored. The goal of involving providers in surveillance measures is to improve HBOC surveillance, which requires wider participation and accurate information.

Strategies

- Include information about family histories and lab test results in state registries.
 - It is possible to model state cancer registries after the data collected by health systems such as Kaiser and Geisinger.
 - A baseline for surveillance could be established using the information given to Myriad with testing orders. States may find it helpful to use this baseline to improve or create the relevant state registry fields.

D. Education

Issues and Challenges

- Awareness of family health history
 - Public health practitioners, officials, and clinicians should be aware of the importance family health history can play in patient care and the national guidelines specifying when to test for HBOC.
 - Clinicians should also be aware of their own educational limits and know when to refer to genetic counseling.
- Lack of genetic literacy
 - The general population does not have an understanding of the details or the likely impact of genetics or of genetic-environmental interaction on their health. Genetics is an up and coming field, and thus there is a deficit in understanding genetics practices and a lack of genetic literacy.
 - It should be up to the state to express the language of policies and practices in simple terms in order to educate the public more effectively.
- Organization and standardization of tools
 - There are several family health history and risk assessment tools available and each has its own strengths and weaknesses. Some tools are designed for use by health care providers, while others are for use by the public. The skill level required by the tools varies because some are intuitive, while others require

training to use. Compiling a list of these resources and organizing them by their target audience would make the tools easier to find and use.

- The family health history and risk assessment tools differ in their focus as well as their target audience. There is a need for standardization of tools to ensure that HBOC diagnosis and screening is included in each of the tools.
- The family health history and risk assessment tools should be easily available to state public health departments.

Strategies

- Family Health History Day
 - Family Health History Day is already in existence and has the potential to make a large impact on awareness of the importance of collecting family health history. Advertising this event could make the public more aware of the importance of family history in HBOC as well as in several other diseases. The increase in awareness of family health history collection could change how patients and doctors approach medical care.
- Improving family health history and risk assessment tools
 - The one problem that exists with collecting family health histories is that once the family has collected the information, there is no clear guideline on what to do with it. Within the directions for the family history tool there needs to be clear direction on where to bring the finished product. Doctors should be educated to know how to integrate the information into patient care.
 - Information should be included about how family members can talk to one another about their family health history. HBOC can be a sensitive topic, especially given different types of relationships within families and between siblings. Acknowledging different family dynamics and diverse conversation styles may help the conversation run more smoothly and successfully.
 - The tools should have some type of feature to easily download to an EMR. Each tool should provide enough direction and education to make patients more informed and prepared for their genetic counseling or doctor's appointments.
- Science curriculum
 - Including genetics in the elementary school science curriculum could have a significant impact on attitudes towards family health histories. Integrating genetics into the science curriculum will likely impact the population in the long term, while also impacting the near future as parents review the information their children bring home from school. The inclusion of genetics in the science standards is currently up for review.
- Partner with relevant advocacy groups
 - It is important to partner with relevant advocacy groups. Using advocacy groups that already have large buy-in from public health departments as well as the general public will allow for quicker dispersal of information and testing resources, in addition to securing buy-in. By partnering public health with these organizations to identify high risk families for HBOC, cascade testing may be more easily performed.

III. Toolkit Recommendations

A. Buy-In

- Recommended Advisory Board Structure
 - A project such as implementing the action plan at the state level will need an advisory board within each state. It may be difficult to put together an advisory board so the toolkit should include suggested roles and give examples.
- Lists of potential partners
 - This list should not simply include names of individuals and organizations, but should also include a description of their work and area of work. Non-traditional potential board members could also be included, such as tumor registrars, patient navigators, public health educators, or accreditation groups. Possible partners are listed below:
 - Laboratories
 - Advocacy groups
 - Pharmaceutical companies
 - Clinicians and health care workers
 - EMR vendors
 - Professional societies
 - Academic institutions
 - Community groups
 - Tumor Board Meeting attendees
 - Tumor registrars
 - Patient navigators
 - Public health navigators
 - Accreditation groups
 - Genetic Science Learning Center representative
- Implementation strategies
 - Include examples from pilot or state programs that have been successful (Michigan and insurance coverage, etc.).

B. Policy

- Summary of current policy
 - The current policies involving HBOC should be stated in medical as well as lay terms, so that they can be easily understood by people of all educational backgrounds.

C. Clinical Intervention

- List of family health history tools
 - Create an organized list of family health history tools, including a description of each tool. The family health history tools could be used for HBOC as well as general use.

D. Surveillance

- Suggested metrics and a database
 - Payer information
 - Patient information
 - Family history
 - Referrals

- The model currently being used in Michigan.

E. Education

- List of available educational resources
 - This should include all new resources that are developed as a part of the action plan.
- Communication plans
 - Possible public service announcements
 - Family Health History Day resources
 - The available resources should be diverse enough to reach different types of people such as providers, families, medical students, etc.
- Provider tools
 - There should be a list available of the tools to identify high-risk individuals, such as the B-RST.

Break-out Session: Lynch Syndrome - Group 1

Facilitator: Kevin FitzGerald, Georgetown University

Notetaker: Lan Le, University of Michigan School of Public Health

Participants:

Frederick Chen, American Academy of Family Physicians

Debra Duquette, Michigan Department of Community Health

Wayne Grody, American College of Medical Genetics and Genomics

Marta Gwinn, Consultant, McKing Consulting Corporation, contractor for Office of Public Health Genomics, CDC

Katherine Johansen Taber, American Medical Association

Laurence Meyer, Salt Lake City VA Healthcare System

Cristi Radford, Lynch Syndrome International

Kathryn Rowley, Utah Department of Health

Albert Terrillion, Consultant

John Tooker, American College of Physicians

Marc Williams, Geisinger Health System

I. Initial buy-in

The group began the break-out session by reviewing the 2009 EGAPP recommendations. Many believed that the recommendation should be updated and expanded to include genetic testing for individuals with endometrial cancer and other LS related cancers. However, in order to expand the current EGAPP recommendations, the CDC must charge the EGAPP Working Group (EWG) with commissioning an updated evidence review through an agency such as the Agency for Healthcare Research and Quality. The importance of having sufficient and appropriate data was a central theme of the discussion; before action can take place, there first must be evidence of cost and lifesaving effectiveness.

A. Action Plan Goal

An action plan for LS screening needs to take a population approach that is substantiated by evidence and metrics. Additionally, any action plan for LS screening should include the caveat that the plan is only applicable for a limited amount of time due to the rapid pace of change in evidence and methodology.

B. Strategies

- The first and most essential step to securing buy-in from public health departments for LS screening is building political will through the identification of effective leader(s) who will excite, connect, and convene the necessary stakeholders. The leader may be a state level champion, someone on the national level or a hybrid. Public health departments can also serve as the leader since they are neutral bodies and may be better equipped to bring together hospitals, providers, consumers and advocacy groups.
- The leader will need to work with the different levels of state leadership. It is important and necessary to bring the state comprehensive cancer programs on board. The CDC's Colorectal Cancer Control Program currently provides funding to 25 states for screening programs that promote population-based CRC screening and follow-up for underinsured or uninsured individuals aged 50-64 years old.²⁰ The leader can point to publications and programs that demonstrate past evidence of success or highlight the cost-effectiveness of LS screening.
- Once the state comprehensive cancer program is on board, it can use its leverage to convene a meeting of state level stakeholders. At the meeting the stakeholders will be asked to define outcomes. Once outcomes have been defined, the state's chronic disease director can be asked to implement data collection and develop informed consent protocols for patients, in addition to protocols for contacting and screening family members.

II. Action Plan Elements

²⁰Division of Cancer Prevention and Control, National Center for Chronic Disease Prevention and Health Promotion (March 1, 2011, updated February 8, 2012). Colorectal Cancer Control Program (CRCCP). Retrieved from <http://www.cdc.gov/cancer/crccp/about.htm>

A. Policy

Issues and Challenges

- According to the EGAPP recommendations there is currently “limited but promising evidence” suggesting that LS screening has clinical utility, improves clinical outcome, and demonstrates cost-effectiveness.²¹ A centralized state database to track compliance and outcomes needs to be developed in order to improve surveillance and inform the development of outcome measures. Defined measures can be used to guide policy development.
- States also need to develop informed consent protocols for MSI and IHC testing as EWG recommends that patient consent be obtained for both screening modalities. These protocols should address when consent should be obtained, who should be responsible for administering consent and what should be included in the consent.
- Related to informed consent for patients, another question that needs to be addressed is who will be responsible for informing relatives if an individual newly diagnosed with CRC tests positive for LS. Protocols for educating, contacting, and testing relatives need to be designed and instituted.

Strategies

- Use examples
 - In the development of individual state policies and action plans, successful LS screening programs from other states can be used as examples to convince state health departments of the potential impact of LS screening for their own state. The CDC can provide examples of projects such as the LSSN, which was started by the Michigan Department of Community Health through funding from the CDC’s Office of Public Health Genomics, as an example of a successful collaborative project. LSSN promotes the implementation of universal screening through the sharing of data and resources such as guidelines and protocols.
 - The publication of research results, tools and guidelines, and evidence based reviews in journals such as *Preventing Chronic Disease* can increase awareness about LS screening among public health practitioners, legislators, and patients.
- Develop partnerships
 - For policy planning and implementation to occur, partnerships need to be developed. In developing partnerships it is important to align the interests of public health departments, academia, payers such as CMS, advocacy groups, professional organizations such as The American Society of Clinical Oncology, American College of Medical Genetics and Genomics, American Gastroenterological Association and American College of Surgeons, testing services companies, and patients.

²¹ Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group. (2009). Recommendations from the EGAPP Working Group: Genetic testing strategies in newly diagnosed individuals with colorectal cancer aimed at reducing morbidity and mortality from Lynch syndrome in relatives. *Genet Med*, 11, 35–41

- The CDC can bring groups together by creating RFPs that require groups such as public health departments and advocacy groups to work together at a state or local level.
- Create a narrative
 - A powerful and persuasive narrative based on the stories of patients is also needed in order to reach both legislators and patients; they may help to elevate awareness of LS to the level currently seen for HBOC. As one group member pointed out, “the development of policy can begin by taking grassroots efforts and bringing them to scale”.
- Create talking points
 - In order to attract the attention of state legislators, talking points limited to two or three bullet points should be developed and disseminated to advocates and those who are likely to lobby for LS screening.
- Caution should be taken with the use of the word “mandate.” In this current environment of health politics, such a charged word may be ill-received by states—especially if funding for a program does not come from a federal entity such as the CDC.

B. Clinical Intervention

Issues and Challenges

- Under the 2009 EGAPP recommendations, LS screening is not universal screening as it is currently limited to MSI or IHC testing of tumor tissue for those newly diagnosed with CRC. There is sufficient evidence to include LS screening for individuals diagnosed with endometrial cancer; however, EWG has not been charged with updating the previous evidence review and recommendation.
- Payers need to expand genetic testing coverage for probands and their relatives. Access to appropriate technology and timely delivery of appropriate care, however, is limited by ICD-9 billing codes.
- The development of metrics and the tracking of outcomes are needed in order to demonstrate the effectiveness of LS screening. Data are needed to evaluate how well providers are recognizing the need to do LS screenings. Only after such data have been collected can policies be implemented to incentivize best practice.

Strategies

- Use EMR
 - Primary clinical transaction and claims data from EMRs are a potential data source.
 - Analysis of these data can help public health departments define and prioritize outcomes for LS screening.
 - In turn, data can be used to lobby CMS to include codes related to treatment and screening for LS and cascade screening for family members in ICD-10.
- Use available resources

- LSSN has a collection of materials such as guidelines, IHC and MSI information sheets, and sample results letters that clinicians and public health departments can use to assist in or enhance clinical implementation of LS screening.
- Competencies for health care providers
 - Core competencies should be established for physician assistants, nurse practitioners, primary care physicians, and specialists.
 - NCHPEG has developed a set of core competencies which emphasizes the need for all health professionals to master knowledge, skills, and attitudes related to genetics/genomics; these competencies can be included in the tool kit as a guide for medical colleges and professional organizations.
- Create incentives
 - Medical societies can incentivize best practices by offering educational programs that meet maintenance of certification requirements, specifically for Part IV (Practice Performance Assessment).
 - To encourage accountability within medical organizations for delivery of care, accountable care organization accreditation can be offered as an incentive.
- Expand infrastructure
 - To ensure access to testing for the underinsured, public health departments should be encouraged to expand their infrastructure to include low cost lab facilities or to work with non-profit labs.

C. Surveillance

Issues and Challenges

- Public access to appropriate information and surveillance data is limited at this time.
- Defined sets of necessary services upon which clinical and public health surveillance can build evidence are needed.
- EMR can be a tool for finding data but adequate funding to hire personnel to conduct surveillance, testing, contact tracing and follow-up is needed.

Strategies

- Clinical surveillance
 - Data agreements with practitioners should be in place so that access to and use of appropriate screening technology such as colonoscopy and trans-vaginal ultrasound, and appropriate care and follow-up visits can be tracked. Evidence of clinical utility and management for patients and relatives would demonstrate how screening can improve health outcomes.
- Public health surveillance
 - Cancer registries should be motivated and encouraged to collect data.
 - Public health departments should also investigate ways to link relatives of those with CRC to the cancer registries.
- Use EMR in both clinical and public health surveillance.
 - EMRs can be used to track family history data and collect statewide data on the number of newly diagnosed CRC individuals who have been screened for LS. In turn, these data can be used to press the case for improving the ICD-10 codes for genetic testing.

D. Education

Strategies

- Educational materials
 - Develop educational materials that are clear, sufficient, and relevant to patients, clinicians, and public health practitioners.
 - For patients, these materials should include guidance on how to talk to their family members about family history, risk, and genetic testing.
- SACGHS and SACHDNC recommendations
 - The group pointed to two reports and recommendations on genetics education and training for health professionals produced by the Secretary's Advisory Committee on Genetics Health and Society (SACGHS) and the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC). These reports can be used as a guideposts for action steps the US Department of Health and Human Services (HHS) can take to integrate and improve genetics education for public health practitioners, point-of-care healthcare professionals, and patients and consumers.
- Educate providers
 - Public health departments can also collaborate with accreditation organizations in order to incentivize education for health care professionals. For physicians, there are resources such as a newly introduced AMA CME course titled “Colorectal Cancer: Is Your Patient at High Risk?” which aims to improve primary care providers’ ability to collect family history, identify patients with increased risk of CRC, and manage their treatment. In addition to being an educational tool, the CME course also acts as an incentive; physicians who complete the online course can receive 6 *AMA PRA Category 1 Credits™*, or if a performance improvement component is completed, 20 *AMA PRA Category 1 Credits™*.

III. Toolkit Recommendations

A. Buy-in

- A collection of evidence based publications
- A collection of narratives
- An inventory of successful programs that have been implemented in other states

B. Policy

- Sample informed consent protocols and screening policies from institutions that have implemented LS screening
- Brief talking points that can be provided to state legislators
- Sample meeting agendas

C. Clinical Interventions

- Lynch Syndrome Screening Guidelines:
<http://www.lynchscreening.net/development/supporting-guidelines/>

D. Surveillance

- Accessible and appropriate data

E. Education

- For everyone: succinct and clear educational materials with relevant information about LS screening procedures and their availability at this time. The educational materials should be one-pagers.
- For physicians: a list of current educational programs for healthcare providers such as AMA's CME course: <https://cme.ama-assn.org/Activity/1068697/Detail.aspx>
- NCHPEG Core Competencies in Genetics For Health Professionals: http://www.nchpeg.org/index.php?option=com_docman&task=cat_view&gid=58&Itemid
- SACGHS Report and Recommendations on Genetics Education and Training: <http://oba.od.nih.gov/oba/sacghs/reports/EducationResolutionJune04.pdf>
- SACHDNC Education and Training Subcommittee Report: http://oba.od.nih.gov/oba/sacghs/study_topics/feb2010/Monaco_ET_Feb2010.pdf

Break-out Session: Lynch Syndrome- Group 2

Facilitator: Debra Lochner Doyle, Washington State Department of Health

Notetaker: Miranda Chergosky, Emory University graduate student

Participants:

Naomi Aronson, Blue Cross and Blue Shield Association

Suzanne Cupal, National Association of County and City Health Officials

W. David Dotson, Office of Public Health Genomics, CDC

Ellen Giarelli, International Society of Nurses in Genetics

Heather Hampel, The Ohio State University

Ira Lubin, CDC

Donald Lyman, National Association of Chronic Disease Directors

James O'Leary, Genetic Alliance

Randi Rycroft, Colorado Department of Public Health

Joan Scott, National Coalition for Health Professional Education in Genetics

Janet Williams, Geisinger Health System

Mary Lou Woodford, Cancer Resource Foundation, Inc.

I. Initial Buy-In

The meeting began with a brief overview of the purpose of the break out session. The use of evidence-based guidelines was emphasized for creating an action plan and toolkit. In the case of LS, this refers to the EGAPP recommendation that all newly diagnosed CRC patients receive screening for LS using either MSI or IHC at the time of diagnosis. Those with an abnormal screening test are offered genetic counseling and testing. When a CRC patient is diagnosed with LS, cascade testing is offered to their at-risk relatives. Participants were strongly encouraged to focus on the current recommended cascade screening strategies for individuals who have already been diagnosed with LS. The entire buy-in and implementation process of the LS action plan relies heavy on the stakeholders and key players.

A. Action Plan Goal

The purpose of the action plan is to implement universal screening in healthcare. This will occur by emphasizing that screening will save lives and cost very little. The development of model policy and legislation language is immediately needed as there is limited time to pass legislation that will be effective by 2014. This includes language at the state level and national level for Medicare. Implementing new policies to pay for LS screening should also be a top priority.

B. Strategies

- Engage relevant stakeholders in surveillance, education, policy, and clinical intervention in the development and implementation of universal LS screening. Relevant stakeholders and champions were defined as healthcare professionals, societies and organizations involved in changing policy. It was noted that many of the organizations would be involved in all four areas, but some will have a more narrow or specific focus. Potential partners to include:
 - Biotech companies, such as Genetech
 - Bayer or other pharmaceutical companies
 - IT support
 - Behavioral, life, and social science researcher groups
 - Lynch Syndrome Screening Network (LSSN)
 - Physicians and physician groups, such as the American Medical Association (AMA)
 - Patient advocacy groups, such as the Genetic Alliance
 - Lobbyists
 - Health educators, such as NCHPEG
 - Healthcare systems
 - Genetic counselors and genetic counseling organizations such as the NSGC
 - Cancer advocacy organizations, such as the American Cancer Society
- Make clear to the different stakeholders that there are cost savings of and benefits to LS cascade screening. It is necessary to show how LS screening will save money in the future.

- Show how lives will be saved using published preliminary data.
- Compile existing state procedures to educate and bring awareness of these policies to all key players within the coalitions and task forces involved in LS screening policies.
- In addition to maintaining support at the state and federal levels, it is important to keep the overall goals of implementing action plans for LS screening “personal” (i.e., real stories of individuals and families that can resonate with all). This focus may be achieved by identifying champions for LS at the state level to push the new policies into state legislation.

II. Action Plan Elements

A. Policy

Issues and Challenges

- There is a lack of data on universal screening that is necessary to direct policy changes. Additionally, the CDC and US Department of Health and Human Services (HHS) do not officially endorse the EGAPP recommendations.
- There is a need to develop academic, federal, state, and private initiatives to generate data and studies to demonstrate and explain to Medicare why cascade screening is necessary. There already are cost-effective data to demonstrate this necessity, but Medicare does not cover cascade screening.
- There is insufficient coverage of all steps in the LS screening process. Problems arise when the patient has coverage for the genetic test, but does not have coverage for the preventative colonoscopy, or vice-versa.
- Medicare should cover genetic testing for unaffected people with a known mutation in their family (cascade testing).

Strategies

- Myriad has previously worked with a regional Medicare group for coverage of some of their tests. Oftentimes, when multiple local service coverage polices are aligned, Centers for Medicare & Medicaid Services (CMS) will adopt the policy as a national standard. If CMS makes a national coverage decision, other payers may follow suit.
- If Medicare does not cover nationally, it is possible to attain similar national coverage through coverage based on each Medicare regional group. However, healthcare reform may make it difficult to cover individuals who have insurance but are unable to pay the deductible, and thus will not qualify for the hardship program.
- Medicare should incentivize universal cascade LS screening.
 - It may be possible to increase the DRG amount for hospitals that perform universal cascade LS screening.
 - Another option would be to make the incentive per patient compensation instead of lowering the DRG amount. Rather than adjust the DRG, institute managed care practices such as offering a per patient per month capitated compensation that would allow the clinicians to provide all medically necessary care including cascade screening for LS after a family member is found to have CRC.

- Create benchmarks and reward hospitals that are providing good care at a low cost with higher reimbursement. These benchmarks may include providing universal screening and demonstrating follow-up of testing results.
- Electronic versions of guidelines for clinical decision-making should be made available to physicians and should be compatible with EMRs.
 - EGAPP should create an electronic version of the guidelines with help from the American Medical Informatics Association (AMAI), Office of the National Coordinator for Health (ONC), and other organizations.
 - Genetic Alliance and Intermountain Healthcare are in the process of organizing a summit that will focus on family health history, genomic information, and EMR. It would be helpful to consider cascade screening for LS during this summit.
- Develop partnerships with industry lobbyists in order to form and advocate for HHS working groups to advise on LS cascade screening. Other partnering groups should include CDC, Health Resources and Services Administration (HRSA), the Personalized Medical Coalition (PMC), National Institutes of Health (NIH) and laboratory organizations to support coverage of testing.
- EGAPP should consider updating their recommendations to address the health benefits to CRC patients identified with LS through universal screening. Many of these patients survive their CRC and if they are diagnosed with LS and receive the appropriate cancer surveillance, we can keep them from developing a second primary LS-associated cancer.
- The Agency for Healthcare Research and Quality (AHRQ) should develop recommendations for surveillance in order to improve the quality, safety, efficiency, and effectiveness of health care.
- Policy changes should include coverage by CMS for genetic counseling sessions.
- Public health professionals should endorse universal LS cascade screening, along with identification of benchmarks and goals. By beginning with public health professionals, it is easier to make policy changes and state mandates for LS screening.

B. Clinical Interventions:

Issues and Challenges

- Many small hospitals do not have genetic counselors. In situations where genetic counselors are not on site, hospitals and their physicians need to provide follow-up for patients who have been screened for LS and appear to be at increased risk. This may be in the form of telemedicine or phone consultation.
- An additional issue is ensuring that those who have screened positive for LS are completing necessary preventive screenings such as annual colonoscopies.
- Genetic counselors need to be reimbursed for their role in coordinating LS cascade testing and/or counseling.
- The clinical outcomes (benefits or harms) for individuals who have undergone cascade screening should be documented.
- In order to help advance clinical implementation, we need studies (and funding of studies) that document improved clinical outcomes of family members diagnosed

- through cascade screening. This will provide further evidence in support of cascade screening.
- Family dynamics may impede LS cascade screening efforts. Patients may not want to contact estranged family members, and strained relationships may lead to ineffective contacts. There will need to be a way to track these unintended negative (and possibly positive) implications of the new program in order to minimize the potential harms.
 - Universal cascade screening for LS in all confirmed CRC patients should be integrated into the practice of pathology.
 - The insurance status of someone with an LS diagnosis affects the quality of care he or she receives for preventive screenings such as colonoscopies. The diagnostic needs of the uninsured should be considered when implementing the action plan.

Strategies

- Develop and use a centralized data warehouse that allows data to be shared among hospitals and is based on a health information exchange design. The data warehouse would be an ideal place to store algorithms for risk assessment and clinical diagnosis. Physicians would have access to information to make better informed decisions for testing, which would make cascade testing more plausible.
- EMR products should include physician reminders and tools to prompt appropriate testing after CRC diagnosis. If reminders are integrated into the system, there would be an additional resource for physicians to utilize to provide optimal care to patients. Additional EMR information categories may include personal and family history as a matrix requirement for the National Committee for Quality Assurance's Patient-Centered Medical Home (NCQA-PCMH).
- Evidence-based guidelines should be developed for referral to appropriate services at point of diagnosis. Patients should also be referred to advocacy groups or family support organizations at the initial point of service.
- Use patient navigators, community health workers and/or case managers to help patients traverse the health system.
- LS screening should be recommended by professional societies.
- Clinicians would be more likely to pursue appropriate cascade screening if they felt confident of being reimbursed for their work. This could be achieved by compiling successful legitimate billing strategies/codes, etc. and advising practitioners about how to use them.
- Create an easy to use decision-making process for the pathologist performing the tests.
- Identify goals for the interventions to better coordinate unified message delivery.

C. Surveillance

Issues and Challenges

- The appropriate use of cancer registries, EMR, and other means of data collection is important for accurate surveillance. Data should be collected through cancer registries on all measures of universal screening for Lynch syndrome, which may include IHC, MSI, *BRAF* mutation, and methylation testing.

- Issues of confidentiality and consent may arise due to the adding of genetic testing results to registry data.
- When implementing LS cascade screening programs, it is necessary to consider diagnosis and treatment follow-up needs of de-identified patients, especially isolated groups like prisoners. For example, when a prisoner has an abnormal IHC result, there is no way to communicate this information to them without special guidance.
- Partnerships are necessary to generate buy-in, and share costs with cancer registries that may be willing to participate in surveillance efforts but lack the financial means to revise their current program or system to capture additional data.

Strategies

- Use cancer registries for tissue banking, family history, and other data acquisitions.
 - Coordinate between state and private registries to achieve standard data elements.
- Use state cancer registries to screen all existing diagnoses of CRC for additional LS testing and possible diagnosis. Analyzing cases within the state cancer registries would close the clinical feedback loop and ensure that patients are not missed.
- The cancer registry should be informed of any data gaps to ensure that problems are minimized.
- Create EMR databases for hospitals to track cases and provide more metrics to justify the implementation of LS screening. EMR systems can implement clinical checks of data to alert clinicians to the need for LS screening, if applicable.
- Coordinate with laboratories for specific data set tracking and metric analysis to gather information about who orders tests, frequency of ordering, and how many tests are performed by the lab each year. This coordination and eventual tracking will be important to measure the success of implementing universal screening.
- A self-sustaining system for LS screening should be created by using clinical and DRG data, in addition to collected data from pilot studies.
- Post-toolkit implementation metrics relating to genetic testing should be monitored to better demonstrate the success and progress of the program. The success of some state programs can also be used by other states to promote national program success.
- Key surveillance partners and organizations include:
 - Surveillance, Epidemiology and End Results (SEER) Program
 - Cancer registries
 - Behavioral Risk Factor Surveillance System (BRFSS)
 - Academic research facilities for LS
 - National organizations of professionals, including nursing organizations

D. Education

The purpose of education from a public health standpoint is to raise awareness and improve skills or change behavior for an improved health outcome. The Health Impact Pyramid places a greater emphasis on public policies since these can impact larger groups as compared to educational efforts that tend to be more targeted and oftentimes

are individually focused as opposed to population based. Preferably, any intervention to alter knowledge, behavior or skills can be measured to determine its effectiveness.

Issues and Challenges

- The challenge with education is to identify which group of potential stakeholders and partners to target first. Education should be considered implementation of the “who, what AND where”, not necessarily just “education.” In other words, the educational strategy must be thoughtful and comprehensive. Universal cascade screening plan education could be directed to consumers, providers, and policy makers.
- Providers’ and the general public’s awareness and knowledge about LS need to be increased. For example, specialists such as pathologists should be targeted with tailored education programs when new algorithms for universal LS cascade screening are developed or new information becomes available.

Strategies

- Tailor existing educational tools using the already existing materials developed by cancer coalitions. The general population is not well informed about LS. For example, many people may not know that CRC can be preventable through colonoscopies.
- Partner with the school systems to add family health history into school projects or have educational days to increase awareness and general knowledge of LS.
- Universal cascade screening for LS is comparable to newborn screening (NBS). The regional counselors for NBS could also counsel for LS.
- Education should extend to appropriate use of laboratory services, i.e., which testing procedure is the best for the patient, whether to do MSI or IHC testing, etc. Pathologist education is necessary to best use limited resources.
- Based on the limited existing funds, funding for testing and education should come from a self-sustaining program. Pilot programs may provide data that can be used in grant or research applications seeking additional funds.
- The CDC can also develop a ‘knowledge network’ for interested states. State health departments that have training programs in place can train neighboring states.
- Review existing literature on culturally specific factors relating to seeking and avoiding genetic testing in various population groups.
 - Partner with relevant community groups and cancer societies to address stigma specific to LS
- Identify the educational needs of each of the different clinicians involved in the universal screening process (i.e., pathologists vs. primary care physicians) and then create tailored education programs.
- In order to educate medical professionals, LS screening should be discussed in grand rounds or at tumor board sessions. During physician education sessions stress why universal cascade screening for LS is necessary and why collecting accurate family health history is important.
- Utilize the media to raise awareness – Popular culture is fascinated by genetic conditions. Perhaps there is a celebrity champion who would be willing to use their status to elevate general awareness of LS.

- Use health communication experts to accurately publicize LS screening through the media
- Utilize patient navigators, community health workers, and advocacy groups to reach out to patients.

III. Toolkit Recommendations

The toolkit should be applicable to all state health departments, regardless of availability and depth of existing local resources and programs. Resources should be categorized and organized for the different audiences who will be accessing them, such as patients, physicians, and public health professionals. It is very important to ultimately implement universal screening for LS in all individuals with confirmed CRC and cascade screening of the relatives of those individuals found to have LS.

A. Buy-in

- Information about other states and existing programs is very important to include in the toolkit. Information contained in the toolkit should include details about that state's champion, the current status of their implementation progress or program success, and what organizations are involved. A map of the fifty states could be used for this progress indicator. When users click on each state in the map, there will be useful links for that state to use, along with different statistics relevant to the genetic epidemiology and screening process, such as the prevalence of LS, and successes and failures in implementing screening. There should also be a "How to" section for states that want to start implementing universal screening.

B. Policy

- Model language for legislation—recommended regulation language.

C. Clinical intervention

- Clinical guidelines for different professionals to use for cascade screening of family members of all individuals identified with CRC.
- Samples of standardized pathology reports.

D. Surveillance

- Links to cancer registries from different states.
- Common data elements for all state cancer registries.

E. Education

- Fact sheets for patients and provider education.
- Information about current statistics for LS.
- Model cases to educate medical professionals about the benefits of universal screening for LS.
- Links to professional organizations and resources like LSSN, AMA, NCHPEG, Genetic Alliance, NSGC, ISONG, etc.

- There should be an educational component of the toolkit which informs laboratories about the proper procedures for notification (i.e., communication for health care providers and families) after testing, including recommendations for genetic counseling as appropriate.
- Social networking could be used to help improve cascade testing and communication in families.

Afternoon Plenary and Concluding Remarks

The afternoon plenary session concluded with summary reports from the facilitators on the goals and strategies for an action plan and the toolkit recommendations discussed in their respective break-out groups. The reports were followed by a brief question and answer session which included the participants, the facilitators, and Dr. Khoury.

In developing a toolkit and action plan for the three Tier 1 applications, Dr. Khoury noted that a common approach can be taken for FH, HBOC, and LS: identify affected individuals, create a narrative, connect patients with services, develop educational materials, develop fact sheets, and provide incentives to providers.

While the need to develop disease specific tools, processes, and infrastructure for HBOC, LS, and FH screening, such as creating centralized registries, developing surveillance measures, defining outcomes, etc., is imperative, Dr. Khoury pointed out that if the three Tier 1 applications are presented to state and local health departments and chronic disease groups as separate agenda items then the strength of the argument for the incorporation, development, and implementation of the genetic screening programs will diminish. When combined together, an argument can be made that nearly two million lives can be saved through the implementation of the three Tier 1 applications by state and local health departments working with clinicians, advocates and other partners.

An effective way to present the action plan to public health departments, state chronic disease directors and patient groups is to create tailored sound bites that emphasize: 1) the early onset and deadly nature of the diseases; 2) the number of lives that can be saved through genetic testing and cascade screening; 3) the effectiveness of the genetic screening intervention in comparison to other disease prevention programs, i.e., salt reduction, smoking cessation etc.; 4) the screening programs' cost-effectiveness ; and 4) the availability of pre-existing resources such as those developed by the LSSN.

Figuring out the feasible next steps that public health departments can immediately take will involve an iterative process. While the three diseases may represent a small fraction of the overall disease burden in each state, the message must get out there that the opportunity to reduce morbidity and mortality through a focus on LS, HBOC, and FH does exist and action should and can be taken now.

APPENDICES

NEW STRATEGIES IN PUBLIC HEALTH GENOMICS: ACTIONS TO SAVE LIVES NOW**Friday, September 7, 2012****Agenda**

- 8:00 am Registration
- 8:30 am Welcome from CDC: Ursula Bauer (Introduced by Muin Khoury)
- 8:40 am Meeting Purpose and Goals: Muin Khoury
- 8:55 am Meeting Logistics: Lan Le
- 9:00 am Lynch Syndrome speaker 1: Heather Hampel
- 9:15 am Lynch Syndrome speaker 2: Debra Duquette
- 9:30 am Lynch Syndrome Q&A
- 9:45 am Hereditary Breast and Ovarian Cancer speaker 1: Mark Robson
- 10:00 am Hereditary Breast and Ovarian Cancer speaker 2: Amber Roche
- 10:15 am Hereditary Breast and Ovarian Cancer Q&A
- 10:30 am Morning break
- 10:50 am Familial hypercholesterolemia speaker 1: James Underberg
- 11:05 am Familial hypercholesterolemia speaker 2: Joan Ware
- 11:20 am Familial hypercholesterolemia Q&A
- 11:35 am Patient and Community Perspectives Panel
Moderator: James O'Leary
- 12:20 pm Break-out session instructions: Toby Citrin
- 12:30 pm Afternoon break and pick up boxed lunch (available for purchase)
- 12:50 pm Break-out discussions: Divide into pre-assigned break-out groups
- 3:30 pm Break and return to auditorium
- 3:45 pm Plenary session: Break-out reports
- 4:20 pm Wrap-up discussion: Karen Greendale and Joan Scott

4:55 pm Concluding remarks: Muin Khoury

5:00 pm Adjournment

Conference Planners

Centers for Disease Control and Prevention, Office of Public Health Genomics

Muin Khoury

Scott Bowen

Karen Greendale

University of Michigan Center for Public Health and Community Genomics

Toby Citrin

Lan Le

Jessica Skiba

Genetic Alliance

James O'Leary

Planning Committee

Sylvia Au, Hawaii State Department of Health

Scott Bowen, CDC/OPHG

Summer Cox, Oregon Health Authority

Rebecca Giles, Utah Department of Health Asthma Program

Karen Greendale, CDC/OPHG

Katherine Johansen Taber, American Medical Association

Jenna McLosky, Michigan Department of Community Health

James O'Leary, Genetic Alliance

Patricia Page, Emory University

Joan Scott, National Coalition for Health Professional Education in Genetics

Amy Zlot, Oregon Health Authority

New Strategies in Public Health Genomics: Actions to Save Lives Now

Conference Participants

Donna Arnett	American Heart Association, arnett@uab.edu
Naomi Aronson	Blue Cross and Blue Shield Association, naomi.aronson@bcbsa.com
Sylvia Au	Hawaii State Department of Health, sylvia@hawaiiogenetics.org
Ursula Bauer	National Center for Chronic Disease Prevention & Health Promotion, CDC, iws8@cdc.gov
Cecelia Bellcross	Emory University School of Medicine, cecelia.a.bellcross@emory.edu
Scott Bowen	Office of Public Health Genomics, CDC, msb4@cdc.gov
Beverly Burke	Connecticut Department of Public Health Genomics Office, beverly.burke@ct.gov
Jean Chabut	Michigan Department of Community Health, chabutj@michigan.gov
Frederick Chen	American Academy of Family Physicians, fchen@u.washington.edu
Toby Citrin	University of Michigan School of Public Health, tcitrin@umich.edu
Wendy Cohn	University of Virginia School of Medicine, wfc2r@virginia.edu
Sara Copeland	HRSA, scopeland@hrsa.gov
Summer Cox	Oregon Health Authority, summer.l.cox@state.or.us
Suzanne Cupal	National Association of County and City Health Officials, scupal@gchd.us
W. David Dotson	Office of Public Health Genomics, CDC, gle1@cdc.gov
Debra Duquette	Michigan Department of Community Health, duquettet@michigan.gov
Lori Farmer	International Society of Nurses in Genetics, dnanurse@att.net
W. Gregory Feero	Dartmouth Medical School, w.gregory.feero@mainegeneral.org
Kevin FitzGerald	Georgetown University School of Medicine, kf3@georgetown.edu
Sabrina Ford	Michigan State University, Sabrina.Ford@hc.msu.edu
Sue Friedman	FORCE: Facing Our Risk of Cancer Empowered, suefriedman@facingourrisk.org
Sandra Fryhofer	AMA Council on Science and Public Health, sandra@drsandrafryhofer.com
Elizabeth Garner	Myriad Genetics, egarner@myriad.com
Ellen Giarelli	International Society of Nurses in Genetics, eg446@drexel.edu
Rebecca Giles	Utah Department of Health Asthma Program, rgiles@utah.gov
Alan Gilstrap	Genzyme, alan.gilstrap@genzyme.com
Karen Greendale	CDC contractor, karengreendale@gmail.com
Wayne Grody	American College of Medical Genetics and Genomics, wgrody@mednet.ucla.edu
Scott Grosse	National Center on Birth Defects and

	Developmental Disabilities, CDC, sgg4@cdc.gov
Marta Gwinn	CDC contractor, mlg1@cdc.gov
Heather Hampel	The Ohio State University, heather.hampel@osumc.edu
Winona Hollins Hauge	Governor's Interagency Council on Health Disparity (Washington State); National Community Council, Genomics SPIG/University of Washington Health Promotion Research Center, whollinshauge@yahoo.com
Yuling Hong	Division for Heart Disease and Stroke Prevention, CDC, ico2@cdc.gov
Paul Hopkins	MEDPED, paul.hopkins@utah.edu
Katherine Johansen Taber	American Medical Association, Katherine.JohansenTaber@ama-assn.org
Muin Khoury	Office of Public Health Genomics, CDC, muk1@cdc.gov
Katherine Kolor	Office of Public Health Genomics, CDC, bxq7@cdc.gov
Stephen Kopecky	Mayo Clinic, kopecky.stephen@mayo.edu
Jane Korn	Minnesota Department of Health, jane.korn@state.mn.us
Lan Le	University of Michigan School of Public Health, lqle@umich.edu
Kimberly Lewis	Georgia Center for Oncology Research and Education, KLewis@georgiacore.org
Debra Lochner Doyle	Washington State Department of Health, debra.lochnerdoyle@doh.wa.gov
Ira Lubin	Public Health Practice Program Office, CDC, ibl0@cdc.gov
Donald Lyman	National Association of Chronic Disease Directors, saclymans@comcast.net
Dana Meaney-Delman	Emory University School of Medicine, dmeaneydelman@gsu.edu
Laurence Meyer	Salt Lake City VA Healthcare System, laurence.meyer@va.gov
Rebecca Nagy	National Society of Genetic Counselors, Rebecca.Nagy@osumc.edu
James O'Leary	Genetic Alliance, joleary@geneticalliance.org
Patricia Page	Emory University, ppage@emory.edu
Cristi Radford	Lynch Syndrome International, cristiradford@gmail.com
Mark Robson	Memorial Sloan-Kettering Cancer Center, robsonm@mskcc.org
Amber Roche	Public Health Seattle & King County, Amber.Roche@kingcounty.gov
Juan Rodriguez	Division of Cancer Prevention and Control, CDC, juan.rodriguez@cdc.hhs.gov
Kathryn Rowley	Utah Department of Health, krowley@utah.gov
Randi Rycroft	Colorado Department of Public Health, randi.rycroft@state.co.us
Debbie Saslow	American Cancer Society, debbie.saslow@cancer.org
Maren Scheuner	VA Greater Los Angeles Healthcare System, maren.scheuner@va.gov
Joan Scott	National Coalition for Health Professional Education in Genetics,

jscott@nchpeg.org

Michael Shapiro
Rochelle Shoretz

Oregon Health & Science University, shapirmi@ohsu.edu
Sharsheret: Your Jewish Community Facing Breast Cancer,
rshoretz@sharsheret.org

Jessica Skiba
Albert Terrillion

University of Michigan School of Public Health, skibaj@umich.edu
Consultant, PHRSLLc@gmail.com

John Tooker
Katrina Trivers

American College of Physicians, jtooker@acponline.org
Division of Cancer Prevention & Control, CDC,
katrina.trivers@cdc.hhs.gov

James Underberg

NYU School of Medicine and NYU Center for Prevention of
Cardiovascular Disease, James.Underberg@nyumc.org

Vickie Venne
Cynthia Vinson

VA Salt Lake City Health Care System, Vickie.Venne@va.gov
National Cancer Institute, cvinson@mail.nih.gov

Joan Ware

National Association of Chronic Disease Directors,
ware@chronicdisease.org

Katherine Wilemon

FH Foundation, katherinewilemon@gmail.com

Janet Williams

Geisinger Health System, jwilliams3@geisinger.edu

Marc Williams

Geisinger Health System, mwilliams1@geisinger.edu

Selvi Williams

Kaiser Permanente Center for Health Research,
selvi.b.williams@gmail.com

Mary Lou Woodford

Cancer Resource Foundation, Inc,
marylou.woodford@cancer1source.org

Barbara Zehnbauer

CDC, ikc8@cdc.gov

Speakers' Biographies

Debra Duquette, MS, CGC has served as the Michigan Department of Community Health (MDCH) genomics coordinator since 2004. She is currently the co-director of a three-year cooperative agreement from the CDC to promote best breast cancer genomics practices. She is also the founder of the Lynch Syndrome Screening Network (LSSN), a consortium of over 90 institutions working to promote universal screening for Lynch syndrome on all newly diagnosed cancers. Ms. Duquette also is the project manager of a CDC Prevention Research Center Special Interest Project to increase breast cancer surveillance and screening for young breast cancer survivors and their at-risk relatives. She is also the project manager for the Michigan Sudden Cardiac Death in the Young (SCDY) Surveillance and Action program. She served as the project manager for a five-year cooperative agreement with CDC to increase genomics in public health programs from 2003-2008 and the project manager for a three-year cooperative agreement with CDC to promote best cancer genomics practices from 2008-2012.

Ms. Duquette received a MS in genetic counseling from Northwestern University in Chicago, Illinois, and a BS in biology and secondary education from Michigan State University. She is a board-certified genetic counselor with over 12 years of clinical genetics experience, providing services to thousands of Michigan families. Her previous places of employment have included Hutzel Hospital in Detroit/SE Michigan from 1993-1998, Sparrow Hospital in Lansing/Mid-Michigan from 1992-1993, and Spectrum Health in Grand Rapids/West Michigan from 1998-2004, where she has been honored to serve and learn from diverse communities within Michigan.

Heather Hampel, MS, CGC is a Professor in the Department of Internal Medicine and Associate Director of the Division of Human Genetics at The Ohio State University. She is the study coordinator for the Columbus-area HNPCC study which enrolled over 1500 colon cancer patients and over 500 endometrial cancer patients to determine the frequency of HNPCC among newly diagnosed patients with these cancers. This study culminated in first author publications in the New England Journal of Medicine in May of 2005, Cancer Research in August of 2006, and the Journal of Clinical Oncology in December of 2008.

Ms. Hampel completed her Bachelor of Science degree in Molecular Genetics at The Ohio State University in 1993. She attained her Master's degree in Human Genetics from Sarah Lawrence College in 1995. She received certification from the American Board of Genetic Counseling in 1996. She worked as a cancer genetic counselor at Memorial Sloan-Kettering Cancer Center in Manhattan before moving to The Ohio State University in 1997. Ms. Hampel was President of the American Board of Genetic Counseling for 2009 and 2010.

Mark Robson, MD is an Associate Attending Physician of the Clinical Genetics and Breast Cancer Medicine Services in the Department of Medicine at Memorial Sloan-Kettering Cancer Center. He is currently the Clinic Director of the Clinical Genetics Service and an Associate Attending of the Clinical Genetics and Breast Cancer Medicine Services. He is also an Associate Professor at Weill Cornell Medical College.

Dr. Robson's research is primarily directed toward improving the integration of genetic information into the clinical management of women with breast cancer. He and his colleagues have conducted a number of studies examining outcomes in women with hereditary breast cancer to better define the risks and benefits of treatments such as breast conserving therapy and adjuvant chemotherapy in this group. He and his coworkers have also conducted a number of studies examining the effectiveness of screening interventions such as breast MRI or ovarian cancer screening in women at hereditary risk. He is currently conducting studies to evaluate the impact of intensive screening or surgical prevention upon women's quality of life, and to develop new screening tools, such as serum peptide profiling. He is also investigating the use of new agents such as PARP inhibitors in the treatment of hereditary breast cancer and the optimal integration of new genetic technologies, such as genomic profiling, into the care of women at risk.

Dr. Robson received his B.Sc. from Washington and Lee University and his M.D. from the University of Virginia. He performed residency and fellowship training at Walter Reed Army Medical Center before coming to Memorial Sloan-Kettering in 1996.

Amber Roche, MPH is the Clinical Preventive Services Manager at Public Health, Seattle & King County (PHSKC). In this role, she oversees the Breast, Cervical, and Colon Health Program (BCCHP) at King County, also serving Kitsap, Clallam, and Jefferson counties. The program helps eligible clients get screened for breast, cervical, and colorectal cancers, and connects clients with diagnostic services and treatment when needed. Eligible clients have low incomes and lack health insurance coverage for cancer screening.

Prior to her position at PHSKC, Ms. Roche was a Health Services Consultant at the Washington State Department of Health, Genetic Services Section. She worked with partners statewide to improve services and follow-up for babies identified with hearing loss through the newborn hearing screening program. She was the liaison to contracted genetics clinics around the state, and participated in the Western States Genetic Services Collaborative. She also partnered with University of Washington researchers on the Genetic Services Policy Project. The GSPP described existing genetic services in the U.S. and developed policy recommendations to address barriers to integrating genetic services into the health care delivery system.

Ms. Roche received her bachelor's degree in biology with a concentration in genetics from Cornell University in 1998, and her MPH in Public Health Genetics from the University of Washington in 2002.

James A. Underberg, MD, MS is a Clinical Assistant Professor of Medicine in the Division of General Internal Medicine at NYU Medical School and the NYU Center for Cardiovascular Disease Prevention. He is the Director of the Bellevue Hospital Primary Care Lipid Management Clinic and is also a member of the executive committee of the Division of General Internal Medicine. His clinical focus is Preventive Cardiovascular Medicine. He is an American Society of Hypertension Certified Specialist in Clinical Hypertension and a Diplomate of the American Board of Clinical Lipidology. Dr. Underberg is president-elect of the Northeast Chapter of the NLA. He also serves on the National Board of the NLA for the term 2011-2014. He is a member of the editorial board of the Journal of Clinical Lipidology, co-chairs the communication committee of the NLA and is the co-editor of the NLA quarterly newsletter Lipid Spin. Dr.

Underberg also serves as the Co-Chair and faculty for the NLA Lipid Academy & Lipid University. He currently serves on the CME committee of the American Society of Hypertension. He is also involved in several clinical trials in the areas of hypertension, lipids, diabetes and cardiovascular disease prevention. He sees patients both in a university based referral practice and in the Bellevue Hospital Lipid Clinic.

Dr. Underberg graduated from Yale University with a BS and MS and from the University of Pennsylvania Medical School. His internship and residency were completed at NYU-Bellevue Hospital Medical Center. He has been elected a fellow of the American College of Preventive Medicine, the Society of Vascular Medicine, the NLA, the American College of Physicians and the American Society of Hypertension.

Joan Ware, BSN, MPH has been active in chronic disease prevention and health promotion for over 30 years. She retired in 2005 from the Utah Department of Health, where she served as Director of the Heart Disease and Stroke Prevention Program. From her first day “on the job” she was involved in strategies to increase awareness of familial predisposition to chronic diseases, including hypertension, FH, and breast and colon cancer. She has worked with leaders in the medical genomics field, including Drs. Roger R. Williams, Paul Hopkins, Ray White, and Mark Scolnick.

Currently, she is a program consultant for the NACDD, partnering with chronic disease directors, CDC’s Divisions of Reproductive Health and Diabetes Translation, state MCH and Diabetes Prevention and Control Program Directors, and the Association of Maternal and Child Health Programs (AMCHP) to promote collaboration and integration of public health services and programs.

Ms. Ware earned a BA in English, a BSN in nursing and a MSPH in Family and Preventive Medicine from the University of Utah.

Panelists' Biographies

Sabrina Ford, PhD is currently involved in cancer education research in the Department of Obstetrics, Gynecology & and Reproductive Biology at Michigan State University. Dr. Ford works specifically with an NIH funded research project entitled the *Kin KeeperSM Cancer Prevention Intervention* that utilizes Community Health Workers to disseminate intense breast and cervical cancer education to Arab, African American and Latina women. The intervention also encourages and tracks breast and cervical cancer screening of the participants. For the last 10 years, she has conducted a number of NIH, federally and locally funded public health projects for underserved populations at the University of Pennsylvania and the Public Health Management Corporation in Philadelphia, PA. Her research expertise involves implementing preventions, interventions, treatment, and outcomes evaluations for vulnerable populations such as children, women, and minorities.

Dr. Ford is a licensed psychologist, health researcher, teacher, and trainer. Dr. Ford received a BS in Psychology from the University of Michigan with a focus on adolescent behavior and a PhD in Counselor Education from the University of Iowa. She completed post-doctoral training at the University of Pennsylvania with a focus on cognitive neuroscience and environmental outcomes. Her background in the study of behavioral factors informs research design to improve health behaviors and promote positive physical health outcomes.

Sue Friedman, DVM was practicing small animal medicine in south Florida in 1996 when she was diagnosed “out of the blue” at age 33 with what appeared to be sporadic breast cancer. At the time, she was unaware of any familial risk factors for hereditary cancer. After her treatment, however, Dr. Friedman realized from an article about hereditary breast cancer that she had several indications for a mutation. She pursued genetic counseling, and in 1997 she tested positive for a *BRCA2* mutation.

Shocked that her health care team didn't alert her to the possibility of being at high risk, and disappointed at having to make critical treatment decisions without knowing of her mutation, Dr. Friedman acted so others could benefit from her misfortune. She founded FORCE in 1999 to fill the information void for individuals and families with hereditary cancer, and to help them advocate for themselves. Under her direction, FORCE has grown into the *de facto* voice of the HBOC community, filling the unique and unmet support needs of those who are navigating risk management and treatment decisions.

With FORCE, no one needs to face HBOC alone. After five years as the organization's executive director and maintaining her own busy practice, Dr. Friedman left veterinary medicine to direct FORCE full-time. Since then, the organization has established itself as an unequalled source of research, advocacy, support, and information regarding risk management, prevention, and awareness. In 2004, Dr. Friedman relocated her family and FORCE headquarters to Tampa to work more closely with researchers to improve options and care for high-risk women.

Winona Hollins Hauge, MSW, LICSW was re-appointed to the Washington State Commission on African American Affairs to serve a second term by the Governor who also requested that she represent the commission on the Governor's Interagency Council for Health Equity. Ms.

Hollins Hauge is the immediate past chair of the UW School of Social Work's Practicum/Field Education Advisory Council. She is currently a member of the UW School of Public Health's Community Advisory Committee where she represents the HPRC on the CDC's National Community Committee. Ms. Hollins Hauge is also on the UW African American Alzheimer's Advisory Council.

Ms. Hollins Hauge joined the Clinical Social Work/BMT team of Fred Hutchinson in 1996, and was promoted to the role of Manager of Community Outreach for Fred Hutchinson Cancer Research in 2002. She completed the five year grant cycle and helped to successfully build and implement a sustainable foundation for the Hutch/Seattle Cancer Care Alliance and partners outreach initiatives. During that tenure she served on the Washington State Comprehensive Cancer Steering Committee, Southwest Oncology Group; National Marrow Donor Programs, ASBMT, Advisory committees, AOSW/BMT SPIG; ACS, and was selected the Washington State representative for the Intercultural Cancer Coalition in her role as Vice Chair of the Washington State Association of Black Health Care Professionals. Ms. Hollins Hauge is currently serving in a private consulting capacity to several national and local organizations who are working on Outreach, Education and Community Partnership building goals.

Ms. Hollins Hauge holds a BA and Masters in Social Work from the University of Washington. She has been the recipient of numerous awards. Her local AOSW peers nominated her for a National Oncology Social Work leadership award. She recently received the Northwest Kidney Center's Community Service Leadership Award and the Seattle Seafair Community Champion Award from the Central Area Senior Centers for excellent service to Elders in the Seattle Community.

Cristi Radford, MS, CGC began her term as President of Lynch Syndrome International (LSI) in June 2012. She is also an education and outreach coordinator at Moffitt Cancer Center. She began her career at Johns Hopkins University in pediatric genetic research and for the last seven years has specialized in cancer genetic counseling in the community setting. In 2011, she implemented the Southwest Florida Lynch Initiative which increased referrals of Lynch Syndrome by over 900%. She is actively involved in fostering collaboration between genetic counselors and nurses and is a columnist for *The Oncology Nurse*.

Ms. Radford received a MS in genetic counseling from the University of South Carolina and a BS in genetics from the University of Georgia.

Rochelle L. Shoretz, JD, is a two-time breast cancer survivor, who founded Sharsheret to connect young Jewish women fighting breast cancer following her own diagnosis at age 28. She is a member of the Advisory Committee on Breast Cancer in Young Women under the auspices of the CDC.

In November 2001, while undergoing chemotherapy treatment, Ms. Shoretz founded Sharsheret, a national not-for-profit organization providing support and resources for young Jewish women facing breast cancer. Since the organization's founding, Sharsheret has launched ten national programs and has responded to more than 25,000 inquiries from those affected by breast cancer, health care professionals, women's and Jewish organizations. Sharsheret's programs and services are open to all women and men, without regard to age, race, religion, or nationality. For its critical services, Sharsheret was awarded the New York

State Innovation in Breast Cancer Research Award and a recent grant from the CDC to develop and launch a national survivorship program for young Jewish breast cancer survivors.

For her pioneering efforts in establishing Sharsheret, Ms. Shoretz was named a “Woman to Watch” by *Jewish Woman Magazine* and a Yoplait Champion in the Fight Against Breast Cancer, and was honored by the Philadelphia Affiliate of Susan G. Komen for the Cure and the Israel Cancer Research Foundation. She is a Board member and graduate of the Joshua Venture Group fellowship of young leaders and a Board member-elect of First Descents, an outdoor adventure program for young cancer survivors. Ms. Shoretz is a graduate of the American Association of Cancer Research Scientist-Survivor Program. As Executive Director and past Board President, Ms. Shoretz has lectured about breast cancer before audiences across the country.

A Centennial Scholar graduate of Barnard College and a Kent Scholar graduate of Columbia Law School, Ms. Shoretz served as a law clerk in 1999 to United States Supreme Court Justice Ruth Bader Ginsburg.

Katherine Wilemon is the founder and president of The FH Foundation. The FH Foundation is the first patient-centered advocacy organization for individuals with FH in the United States.

Under Ms. Wilemon’s leadership, the FH Foundation has gained the support and commitment of top lipidologists from across the country to guide the launch of a National Familial Hypercholesterolemia Registry. Another key initiative of the FH Foundation is the observance of the first National FH Awareness Day on September 20, 2012. Ms. Wilemon is also a board member of the International FH Foundation. Ms. Wilemon works full-time in a volunteer capacity to raise awareness of FH and increase the rate of accurate diagnosis and proactive treatment. She was herself diagnosed with FH only after having a heart attack at age 39.

Ms. Wilemon received her B.S. in Behavioral Medicine and Psychology summa cum laude from the University of North Florida.