Recommendations for a Public Health Approach to Cancer Genetics/Genomics in Pennsylvania

A Report from the Pennsylvania Cancer Control, Prevention and Research Advisory Board (CAB) to the Pennsylvania Secretary of Health

March 2019

pennsylvania
DEPARTMENT OF HEALTH

March 4, 2019

Rachel Levine, M.D.
Secretary of Health
Pennsylvania Department of Health

Dear Dr. Levine:

Following the charge from Dr. Loren Robinson, Deputy Secretary for Health Promotion and Disease Prevention, the Pennsylvania Cancer Control, Prevention and Research Advisory Board (CAB) formed the Cancer Genetics/Genomics Committee to provide recommendations on a statewide public health approach for Cancer Genetics/Genomics to be included in the *2019-2023 Pennsylvania Cancer Control Plan*.

We hereby submit to you our report of recommendations for a Public Health Approach to Cancer Genetics/Genomics in Pennsylvania. This report addresses public health promotion and disease prevention utilizing recent and emerging knowledge in the field of Cancer Genetics/Genomics. The recommendations focus primarily on data collection and surveillance to monitor the burden of hereditary cancers, education of the population and providers regarding genetic predisposition to cancer, genetic counseling, appropriate testing, screening and other risk management services, and promotion of policy and systemic change to advance Cancer Genetics/Genomics in Pennsylvania to save lives and improve the health and quality of life of Pennsylvanians. This issue is timely and of utmost importance, since the Commonwealth lags behind more than 60% of states in adding Genetics/Genomics Goals to our Cancer Plan.

Genetics/Genomics information is integral to early cancer diagnosis, treatment, and consequent reduced cancer morbidity and mortality; momentum should not end with this report. One recommendation of the Committee is the formation of a Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health, initially to oversee and manage the implementation of this report, including pressing health policy and legal issues that go beyond cancer control and prevention.

Members of the Cancer Genetics/Genomics Committee have voiced strong interest and commitment to ensure implementation of the recommendations within the report. We propose formation of a Cancer Genetics/Genomics Workgroup of the Pennsylvania Cancer

Coalition to undertake this important mission and work collaboratively with the proposed Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health.

We look forward to the opportunity to discuss this report and its recommendations with you.

Sincerely yours,

Margaret O'Grady, MSN, OCN, FAAMA
Vice Chair, Pennsylvania Cancer Control, Prevention and Research Advisory Board

Susanne M. Gollin, Ph.D., FFACMG
Member, Pennsylvania Cancer Control, Prevention and Research Advisory Board

Executive Summary

The Pennsylvania Cancer Control, Prevention, and Research Advisory Board (CAB) Cancer Genetics/Genomics Committee is submitting this Public Health Approach to Cancer Genetics/Genomics in Pennsylvania report to the Pennsylvania Secretary of Health. The report highlights the importance of surveillance to monitor the burden of hereditary cancers, education to increase awareness amongst the public about genetic predisposition to cancer, and amongst providers about genetic counseling, testing and risk management services, and promotion of policy and systemic change to advance Cancer Genetics/Genomics in Pennsylvania to save lives and improve health and quality of life. This report addresses the need to have a comprehensive approach in Pennsylvania to address Cancer Genetics/Genomics.

The strategic objectives of this report are to:

- Assess the burden of hereditary cancers for individuals, families, and communities, and the use of genetic counseling, genetic testing and associated clinical services.
- Increase the knowledge amongst the public and health care providers about hereditary cancers, genetic counseling, appropriate genetic testing, and associated clinical services.
- Improve access to and insurance coverage of genetic counseling, genetic testing, and associated services for high-risk individuals and families.
- Facilitate implementation of the recommendations in this report by building Public Health Genetics/Genomics infrastructure in the Commonwealth of Pennsylvania.

The report begins with the burden of hereditary cancers, and is organized around the areas comprising a comprehensive Public Health Approach to Cancer Genetics/Genomics in Pennsylvania: Data and Surveillance, Education, and Policy, Insurance and Systemic Change.

The recommendations in support of Public Health Cancer Genetics/Genomics in Pennsylvania are:

For Data and Surveillance:

1. **Report cancer genetics/genomics data and health disparity data by:**
   a. Enhancing the Behavioral Risk Factor Surveillance System (BRFSS) questionnaire to include several questions about access and uptake of genetic counseling and/or testing, awareness and referral rates.
   b. To incorporate genetic testing information into the state cancer registry.
   c. Assessing available cancer registry data that relate to cancer genetics and genomics to determine future uses of the data (i.e., bi-directional cancer registry reporting).
   d. Establishing state level data from the Healthy People 2020 national data.
   e. Investigating Medicaid claims data related to genetic testing.
2. **Create a Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health** to coordinate implementation of the recommendations of this report, advance the Genetics/Genomics goals of the Cancer Control Plan, and promote awareness of and manage programming to address genetics/genomics issues in Pennsylvania.

3. **Increase access to genetic services in underserved areas in Pennsylvania** by identifying geographic disparities in service, supporting alternate service delivery models for genetics services, identifying funding options to improve access, and pursuing partnerships with State, Regional, and National organizations working towards similar goals (i.e., CDC and the NYMAC Regional Genetics Network).

For Education:

1. **Develop a Workgroup in the Pennsylvania Cancer Coalition** to implement recommendations from this report regarding Cancer Genetics/Genomics Education, Data and Surveillance, and Policy, Insurance Coverage, and Systemic Change.
2. **Provide ongoing and updated provider education** related to 1) identification and appropriate referrals for at-risk patients, 2) cancer risks related to genetic syndromes, 3) follow up screening/medical management guidelines, and 4) cancer treatment implications associated with hereditary cancer risks. These resources can come in the form of:
    a. A state-supported webpage providing links to testing guidelines, referral resources, and other relevant topics, e.g., links to National Comprehensive Cancer Network (NCCN), US Preventative Services Task Force (USPSTF), and other guidelines and searchable provider databases, e.g. www.findageneticcounselor.com.
    b. Development of simplified "guides" to assist providers in identifying at-risk patients in their clinics for referrals for genetic counseling/testing.
    c. Web-based CME/CNE training program, e.g. webinars.
    d. In-person training sessions at health centers throughout the state, provided by trained educators and a standardized slide set.
3. **Provide ongoing and updated public education related to hereditary cancer risk factors and how to find cancer genetics specialists**. These resources can come in the form of public service announcements and a State-supported webpage providing links to testing guidelines, referral resources, e.g., www.findageneticcounselor.com, a statewide provider database, and other relevant topics.

For Policy, Insurance and Systemic Change:

1. Facilitate legislation to **create a public health genetics/genomics infrastructure** in the Commonwealth of Pennsylvania.
2. Support the proposed **amendment to the genetic counselor licensure law** to allow genetic counselors to order tests.
3. **Seek funding** for a Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health to oversee and manage the implementation of this report, to incorporate genetic testing information into the state cancer registry and add cancer genetic testing and services questions in the BRFSS.

These recommendations have been developed to guide the inclusion of genetics and genomics goals in the *2019-2023 Pennsylvania Cancer Control Plan*. The members of the Genetics/Genomics Committee look forward to the ongoing collaboration with the CAB, the Pennsylvania Department of Health, the PA Cancer Coalition, and numerous states, regional

and national stakeholders in implementing the recommendations outlined in this report to improve cancer prevention and care for Pennsylvanians.

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Cancer Genetics/Genomics Committee Members

Chairs:

Susanne M. Gollin, PhD, FFACMG is a Professor of Human Genetics at the University of Pittsburgh Graduate School of Public Health and served as a Clinical Cancer Cytogenetics Consultant in the Pittsburgh Cytogenetics Laboratory of the University of Pittsburgh Medical Center (UPMC). Since earning her Ph.D. at Northwestern University and training in Clinical Cytogenetics at Baylor College of Medicine, Susanne has had a long and fulfilling career as a cancer genetics researcher, educator, mentor, innovator, and clinical cancer cytogeneticist, board certified by and a Founding Fellow of the American College of Medical Genetics and Genomics. Susanne has served as a Consultant and Member of the Medical Devices Advisory Committee of the U.S. Food and Drug Administration, a Member of the Clinical Laboratory Improvement Advisory Committee, Proficiency Testing and Genetic Testing Subcommittees at the U.S. Centers for Disease Control and Prevention, a member of the Pennsylvania Cancer Control, Prevention and Research Advisory Board, and the editorial boards of *Genes, Chromosomes & Cancer*, *Cytogenetic & Genome Research,* and *Melanoma Research*, amongst other scholarly journals. Her research interests include cytogenetic alterations as diagnostic, prognostic, and therapeutic biomarkers for cancers; mechanisms involved in chromosomal instability; and defects in the DNA damage response in cancer cells and their role in therapeutic resistance. A breast cancer survivor, Susanne is on a mission to apply knowledge of genetics and genomics to make cancer a chronic disease, rather than a deadly one.

Margaret A. (Peg) O'Grady, MSN, OCN, FAAMA is currently the Administrative Director of the Abington-Jefferson Health System's Asplundh Cancer Pavilion. She overseas inpatient and outpatient oncology service including a robust research relationship with the Sydney Kimmel Cancer Center. Peg was previously the Director of Nursing for the Sydney Kimmel Cancer Center Medical Oncology division. She has significant expertise in oncology care coordination also having worked at Fox Chase Cancer Center as the Senior Director of the first cancer center network in the United States – The Fox Chase Partners Program supporting development of 30 plus institutions cancer centers. She is currently the Vice Chair of the Pennsylvania Cancer Control, Prevention, and Research Advisory Board. She is the past President of the Pennsylvania Society of Oncology and Hematology, the state-wide American Society of Clinical Oncology group, and is the past President of the American Academy of Medical Administrators. Her research interests are in health outcomes, transition of care and navigation having published in breast and colorectal navigation processes.

Members:

David Buono, Jr. is a Consumer Liaison with the Pennsylvania Insurance Department. With experience working in the insurance industry, David now leads the Insurance Department's increased efforts to provide consumer outreach and communications. His primary role is to make sure the department is reaching consumers and helping them navigate the complex world of insurance. He supports the department's initiatives to keep the state's insurance market competitive, while focusing on helping consumers learn and understand their rights and

responsibilities, what options are available to them when it comes to insurance products and providing a place for consumers to get answers to their questions.

Susan Domchek, MD is the Basser Professor in Oncology at the University of Pennsylvania. She is the Executive Director of the Basser Center for BRCA at the Abramson Cancer Center. Dr. Domchek is a medical oncologist with expertise in breast and ovarian cancer risk assessment, genetic testing, and management of individuals with inherited cancer susceptibility. Dr. Domchek has published more than 250 articles in this field. She serves on the editorial board for the *Journal of Clinical Oncology* and on the Scientific Advisory Board for the Breast Cancer Research Foundation. Dr. Domchek is an elected member of the National Academy of Medicine.

Andrea L. Durst, MS, DrPH, LCGC is a certified and licensed genetic counselor with 8 years of experience in cancer genetic counseling. She is currently the Associate Director of the Genetic Counseling Program and the Co-Director of the MPH Program in Public Health Genetics at the University of Pittsburgh where she teaches courses on both genetic counseling and public health genetics. She received her Master of Science in Genetic Counseling from the University of North Carolina at Greensboro and her Doctor of Public Health in Health Management and Policy from the University of Kentucky where she investigated the potential for identification of individuals appropriate for genetic referral via bidirectional cancer registry reporting. Dr. Durst is currently working with several organizations to advance public health genetics projects. She serves as Chair of the Steering Committee for the New York Mid-Atlantic Regional Genetics Network (NYMAC), as Secretary/Treasurer of the Pennsylvania Association of Genetic Counselors (PAGC), and previously served as Co-Chair of the National Society of Genetic Counselors (NSGC) Public Health Special Interest Group. Dr. Durst previously provided her public health genetics management expertise to the Midwest (Region 4) Genetics Network and The Genetic Alliance.

Andrea Forman, MS, LCGC is currently the Senior Genetic Counselor and Clinical Team Lead at Fox Chase Cancer Center in Philadelphia. She received her master's degree in Genetic Counseling from the Mount Sinai School of Medicine in New York City in 2004. She spent four years working with the Department of Cancer Prevention and Control at Mount Sinai, coordinating research studies on innovative genetic counseling techniques, such as culturally tailored genetic counseling for women of African Ancestry. She joined the Department of Clinical Genetics at Fox Chase Cancer Center in 2008. As part of her work at Fox Chase, she provides genetic counseling services for all different cancer types, supports research studies on tumor and germline genomic testing and other topics, supervises and mentors genetic counseling trainees, and gives educational lectures to medical professionals and students in the Philadelphia region. She recently completed a 2-year role as co-chair of the Cancer Special Interest Group for the National Society of Genetic Counselors.

Veda N. Giri, MD is an Associate Professor in Medical Oncology and Cancer Biology at Sidney Kimmel Cancer Center at Thomas Jefferson University. She is a medical oncologist with specialization in clinical cancer genetics. Dr. Giri is Director of Cancer Risk Assessment and Clinical Cancer Genetics at Thomas Jefferson University, where she leads an integrated and comprehensive effort in inherited cancer risk assessment and conducts studies focused on

genetic evaluation of cancer risk. She has focused interest in developing the field of genetic counseling and genetic testing for inherited prostate cancer. Dr. Giri started the first Men's Genetic Risk Clinic in the US in 2014 focused on genetic evaluation of inherited prostate cancer in the setting of multidisciplinary care. Dr. Giri also co-chaired an international consensus conference that published a comprehensive framework for genetic evaluation of prostate cancer. She is the PI of the Genetic Evaluation of Men (GEM) study, which has contributed insights into the germline spectrum of men undergoing multigene testing for inherited prostate cancer, as well as insights into the needs of men undergoing genetic counseling. Dr. Giri received her medical degree from Jefferson Medical College (now Sidney Kimmel Medical College) and completed her residency in Internal Medicine and fellowship in Hematology-Oncology at the University of Michigan. She then completed advanced training in molecular cancer genetics and cancer risk assessment at Fox Chase Cancer Center. Dr. Giri has served on national committees, including the National Comprehensive Cancer Network Prostate Cancer Early Detection Panel and NIH PDQ® Cancer Genetics Editorial Board, contributing expertise in cancer risk assessment and prostate cancer genetics.

Jennifer M. Johnson, MD, PhD completed her undergraduate degrees in Biology and Theology at St. Joseph's University before moving to the University of Pittsburgh where she obtained both her MD and a PhD in molecular genetics, studying DNA damage and repair in breast cancer. She completed her clinical training at Thomas Jefferson University Hospital (TJUH) as a resident and chief resident in Internal Medicine followed by a fellowship in Hematology and Medical Oncology. She currently serves on the faculty at TJUH as an Assistant Professor and is the Associate Program Director for the Hematology and Medical Oncology Fellowship. Her interest in medical education and background in molecular genetics has manifested in the creation of an enterprise-wide Molecular Tumor Board, which connects four hospitals within the Jefferson system in an online educationally-focused case-based discussion. Dr. Johnson's current research interests lie in translational medicine in aerodigestive cancers, focusing on the tumor microenvironment from both a metabolic and immunologic standpoint. Her collaborations with benchtop researchers at Jefferson have led to multiple clinical trials ongoing within the fields of head and neck cancer and lung cancer, both to gather further information on the ways in which cancer cells work together with their non-cancerous supportive cells, and to test new therapeutic options to disrupt this critical interaction. While looking ahead to these next therapeutic horizons, she has also been involved in expanding supportive care for our patients with enhanced supportive care including the use of technology to improve patients' ability to record and quantify treatment-related side effects.

Monika Joshi, MD, MRCP is an Associate Professor in the Division of Hematology-Oncology, Department of Medicine, at the Penn State Cancer Institute (PSCI). She specializes in genitourinary (GU) malignancies. She has expertise in the development of clinical trials with Immunotherapy based therapeutic combinations. She is the principal investigator on a number of clinical studies and has authored publications in a number of peer-reviewed journals in oncology. She is the GU disease team Co- leader at the PSCI. She also serves as director for the Molecular Tumor Board at PSCI and the Co-chair for GU committee for the Big Ten Cancer Research Consortium. She is an active member of the GU core committee for ECOG-ACRIN

consortium and is involved in clinical trial development. Her research interests include development of clinical trials in GU cancer, predictive and prognostic genomic biomarkers for bladder cancer; role of DNA damage response and repair genes in response to therapy in bladder cancer.

David H. Ledbetter, PhD is executive vice president and chief scientific officer at Geisinger. Previously, he held academic and leadership positions at Emory University, the University of Chicago, and the National Center for Human Genome Research at NIH. He is a graduate of Tulane University and earned his Ph.D. at the University of Texas-Austin. He is an internationally recognized expert in Genomics and Precision Medicine, having focused his early research efforts on discovering the genetic causes of childhood neurodevelopmental disorders such as autism, and the translation of new genomics technologies into clinically useful genetic tests for early diagnosis and intervention. His current research interest includes leveraging longitudinal electronic health information with large-scale DNA sequencing to determine the clinical utility and cost-effectiveness of precision medicine approaches in a real-world health system setting.

Phuong L. Mai, MD, MS is an Associate Professor in the Department of Obstetrics, Gynecology and Reproductive Sciences, University of Pittsburgh School of Medicine and is the Medical Director of the UPMC Cancer Genetics Program. Prior to joining the University of Pittsburgh School of Medicine, she was a staff clinician in the Clinical Genetics Branch of the National Cancer Institute's Division of Cancer Epidemiology and Genetics, where she was involved in a number of epidemiologic and clinical research studies aimed at better understanding familial cancer syndromes, particularly cancer risks and early detection and interventions in Hereditary Breast Ovarian Cancer Syndrome and Li-Fraumeni Syndrome. Since joining the University of Pittsburgh, she has been working to expand cancer genetics services, especially to populations in rural areas via telemedicine. Dr. Mai is also a member of the NRG Oncology's Cancer Prevention and Control Committee and is on the Medical Advisory Board of the Li-Fraumeni Syndrome Association (LFSA), a patient advocacy group serving the LFS family community. Her clinical and research work focuses on hereditary cancer risk assessment and management, and the delivery of cancer genetic services to underserved populations.

Khadijah A. Mitchell, PhD, MS is an Assistant Professor of Biology at Lafayette College and the Principal Investigator of the Integrative and Translational Laboratory for Applied Biology (IT LAB). She researches the causes and consequences of cancer health disparities in vulnerable populations and teaches Precision Medicine and Molecular Genetics. Prior to joining Lafayette, Dr. Mitchell worked at the National Cancer Institute and taught at the FAES Graduate School at the NIH. She has more than 15 years of experience in laboratory research, public health practice, higher education leadership, and government service. As a scholar-teacher-advocate, Dr. Mitchell's work has appeared in high-impact research journals, contributed to programs that reduce health disparities in underserved populations through community engagement, and championed the recruitment and retention of women and minorities in STEM careers. Dr. Mitchell earned her Ph.D. in Human Genetics and Molecular Biology from the Johns Hopkins School of Medicine. She concurrently earned a graduate certificate in Health Disparities &

Health Inequality from the Johns Hopkins Bloomberg School of Public Health, and was a trainee in the Johns Hopkins Center to Reduce Cancer Disparities. Previously, she earned her M.S. in Biology from Duquesne University and B.S. in Biology from the University of Pittsburgh.

Diane Phillips is the Director of Government Relations for the American Cancer Society Cancer Action Network (ACS CAN) in Pennsylvania. Since 2000, she has worked with volunteers, staff, coalitions and public officials to promote state policies that address cancer prevention, early detection, research and treatment. Working with coalition partners, she led a successful campaign to increase tobacco taxes during the 2015-16 state legislative session. As a result, the per pack tax on cigarettes was increased by $1.00 in July 2016 and other tobacco products are now taxed for the first time. She also helped to successfully lobby for the inclusion of tobacco prevention and cessation programs in Pennsylvania's Master Settlement Agreement plan in 2001 and is currently working with coalition partners to strengthen the state's Clean Indoor Air Act. Diane was also part of the coalition that worked to pass the commonwealth's smoke-free law in 2008. Prior to joining the Society, Diane worked for 10 years in legislative advocacy and community programs with the American Heart Association during 1990-2000. She was part of the original tri-agency coalition of Heart, Lung and Cancer that formed in 1998 to address Pennsylvania's tobacco settlement distribution plan. She was also successful in extending Good Samaritan coverage for use of automated external defibrillators (AEDs) in Pennsylvania when they were first made available to the public. Diane has also served as the Executive Director for the ARC in York County, and as a statewide community specialist for the ARC's Pennsylvania office. The ARC provides advocacy and developmental services for persons with intellectual disabilities. In addition, she served as a planner for the regional health systems agency in south central Pennsylvania. Diane holds a bachelor's degree in international studies from American University in Washington, DC and has completed graduate course work in public administration at the Pennsylvania State University, Harrisburg.

Alanna Kulchak Rahm, MS, PhD, LGC is a researcher in the Geisinger Genomic Medicine Institute. She is a licensed genetic counselor and holds a doctorate in Health and Behavioral Science. She has over 20 years' experience in cancer genetics and currently serves on the national Lynch Syndrome Screening Network (LSSN) Board of Directors. Her personal research focuses on the implementation of genomics to improve population health; including implementation of Universal Lynch syndrome screening, cascade genetic testing, patient and provider understanding of genomics, and patient engagement in research. She has conducted public health and health systems research for nearly two decades, has collaborated on initiatives to improve genomics education in Native Americans, has conducted research on understanding barriers to cancer genetic testing, and conducted research on provider education and understanding of genomics. She is currently the principal investigator of an NCI-Beau Biden Cancer Moonshot-funded project to study the implementation of universal tumor testing for Lynch syndrome across multiple healthcare systems, collaborates on studies to implement family health history assessment in healthcare systems, and leads a study to implement breast cancer risk assessment on all women at screening mammography in Geisinger.

Margery Wasko, MD graduated from Mount Holyoke College in Massachusetts, received her MD from Columbia University College of Physicians and Surgeons in New York City, and trained in Pediatrics at Rainbow Babies and Children's Hospital and Cleveland MetroGeneral Hospital of Case Western Reserve University in Cleveland, Ohio. She is Board Certified in Pediatrics and practiced for 23 years at Federally Qualified Health Centers, including 5 years in Cleveland and 18 years at Hamilton Health Center in Harrisburg, PA. Since 2007, she has worked as a Consultant Medical Director in the Office of Medical Assistance Programs (Bureau of Fee For Service Programs) of the Pennsylvania Dept. of Human Services, with the last 3.5 years as the Lead Consultant Medical Director. Dr. Wasko was on the medical staff of PinnacleHealth for many years, where she served as Chair of the Dept. of Pediatrics QA Committee and is now serving her 24th year on Pinnacle's Institutional Review Board. She also was the Pennsylvania Chapter of the American Academy of Pediatrics' representative to the state Medical Assistance Advisory Committee (MAAC), and she currently volunteers as a member of the Board of the Community Check-up Center in Harrisburg.

Janet L. Williams, MS has held a variety of genetic counseling positions in multiple locations across the U.S. including Wisconsin, Utah, California and Pennsylvania. For the past 25 years, Janet has provided genetic counseling in prenatal, pediatrics, cancer and adult clinical settings. She is now an Assistant Professor in Genomic Medicine Institute at Geisinger in Danville, PA. Janet's research interests include defining outcomes related to genetics service delivery, in particular outcomes related to genetic counseling encounters. Janet is involved in a local research study utilizing whole genome sequencing in children with undiagnosed Intellectual Disability, Autism Spectrum Disorder and/or congenital anomalies. Ms. Williams completed research funded through the Patient Centered Outcomes Research Institute (PCORI) which developed online genome sequencing results reports for patients and providers. Through parent and provider engagement, reports were developed that enhanced communication and shared decision making for parents and providers.

Pennsylvania Department of Health

Wendy Aldinger, RHIA, CTR is the Manager of the Pennsylvania Cancer Registry (PCR). As manager she oversees the overall operations of the PCR by monitoring the compliance with CDC National Program of Cancer Registry (NPCR) standards. Ms. Aldinger represents the PCR on the Pennsylvania Cancer Advisory Board, the Pennsylvania Cancer Coalition, Data Advisory Committee, Pennsylvania Cancer Control Leadership Team and the Public Health Gateway Governance Program and Project Committee.

April Barry, LSW, MSW is the Performance Improvement and Evaluation Manager for the Comprehensive Cancer Control Program. In this position, she is responsible for providing leadership and strategic direction to assess, plan, and implement performance improvement/evaluation for comprehensive cancer control and oversee implementation of cancer initiatives. She serves as co-chair of the National Colorectal Cancer Roundtable Evaluation and Measurements Task Group and serves on the Pennsylvania Department of Health Quality Improvement/Performance Management Council. She holds a master's in social work from Temple University, is a Licensed Social Worker in Pennsylvania, holds an

undergraduate degree from Elizabethtown College, holds certifications in Patient Navigator and Performance Improvement Practice Facilitation and has more than 28 years public health and healthcare experience.

Sharon Sowers is the Director of the statewide Comprehensive Cancer Control Program**.** She has more than 30 years of health care experience having worked in regional health systems, a professional association, managed care organization, long-term care institutions, and both insurance and state government. Her specialization is in marketing, public/provider relations, planning board/coalition management, program administration and consulting. Sharon and her team develop, implement and evaluate cancer control strategies.

The CAB Cancer Genetics/Genomics Committee Workgroups

Report Introduction: Andrea L. Durst, MS, DrPH, LCGC and Veda N. Giri, MD.

Data and Surveillance: Andrea L. Durst, MS, DrPH (lead), Wendy Aldinger, RHIA, CTR, Susanne M. Gollin, Ph.D., FFACMG, and Monika Joshi, MD, MRCP.

Education: Andrea Forman, MS, LCGC (lead), Susan Domchek, MD, Veda N. Giri, MD, Jennifer M. Johnson, MD, PhD, Phuong L. Mai, MD, MS, Khadijah A. Mitchell, PhD, MS, and Margaret A. (Peg) O'Grady, MSN, OCN, FAAMA.

Policy, Insurance and Systemic Change: Alanna Kulchak Rahm, MS, PhD, LGC (lead), David Buono, Jr., Diane Phillips, Margery Wasko, MD, and Janet L. Williams, MS.

Public Health Approach to Cancer Genetics/Genomics in Pennsylvania
(Cancer Genetics/Genomics Committee Report/Recommendations)

Introduction

Through the leadership of the Pennsylvania Cancer Control, Prevention, and Research Advisory Board (CAB), the Pennsylvania Cancer Coalition and the Pennsylvania Department of Health is currently developing the *2019-2023 Pennsylvania Cancer Control Plan* to serve as a guide for cancer prevention and management activities in the Commonwealth. Over the years, these cancer control plans have focused on high-impact goals, including improving health disparities, patient navigation, and access to affordable health care, amongst others. The CAB addresses both timely and cutting-edge issues in cancer prevention, control, and care of the population of Pennsylvania by development of a new cancer plan every five years.

In 2018, the Pennsylvania Department of Health tasked the CAB with developing recommendations for advancing cancer genetics/genomics in Pennsylvania to assess the burden of hereditary cancers, educate the public and health care providers about genetic predisposition to cancer, and facilitate genetic counseling, appropriate testing, screening, and appropriate risk management through surveillance and risk-reducing interventions, leading to early diagnosis and reduced cancer morbidity and mortality. Whereas more than 60% of states (36/51, including the District of Columbia) included at least one goal related to cancer genetics/genomics in their most recent Cancer Control Plan [1], Pennsylvania was not one of these states. In 2012, the Centers for Disease Control and Prevention (CDC) Office of Public Health Genomics (OPHG) established an evidence-based classification system for the readiness of genomic applications in practice. Tier 1 applications, which are considered ready for implementation, include two hereditary cancer syndromes, Hereditary Breast and Ovarian Cancer syndrome (HBOC) specifically due to pathogenic variants (mutations) in the *BRCA1* and *BRCA2* genes and Lynch syndrome (LS) [2-4]. More recently, the CDC OPHG has recognized the vital role of state public health departments in successfully executing these Tier 1 applications [3, 5]. The increased focus of state and federal public health agencies on cancer genetics/genomics and the importance of identifying individuals in Pennsylvania at significantly increased risk for cancer due to these two cancer predisposition syndromes serve as the motivating factors for the formation of the Cancer Genetics/Genomics Committee and adding genetics/genomics goals to the *2019-2023 Pennsylvania Cancer Control Plan*.

Approximately 10% of breast cancers and as many as 24% of ovarian cancers are due to alterations in genes associated with HBOC [6-8]. The prevalence of alterations in the two most common genes associated with HBOC, *BRCA1* and *BRCA2*, has been estimated to be approximately 1:400 to 1:500 [9, 10], although several analyses, including a recent study completed at a major health system in Pennsylvania, provides evidence that the prevalence of alterations in these two genes may be substantially higher than previously thought, up to 1:180 [11, 12]. Based on this information, it is estimated that approximately 25,600-71,100

Pennsylvanians carry a pathogenic variant in *BRCA1* or *BRCA2*, many of whom do not know they are at an increased risk of cancer (Table 1) [13, 14].

Individuals with LS are at increased risk of developing colorectal, endometrial, ovarian, gastric, and other cancers. As many as 3% of colorectal cancers and 1.8% of endometrial cancers are due to alterations in the five genes associated with LS (*MLH1, MSH2, MSH6, PMS2* and *EPCAM*) [15-17]. The prevalence of alterations in these genes is estimated to be approximately 1:279 [18], meaning that approximately 45,900 Pennsylvanians carry a pathogenic variant in one of the genes associated with LS (Table 1).

Table 1: Prevalence of HBOC/LS and Estimated Numbers of At-Risk Individuals in Pennsylvania

	Prevalence[a]	Estimated Number of Pennsylvanians At-Risk[b]
Hereditary Breast & Ovarian Cancer Syndrome (*BRCA1* and *BRCA2* genes)	1:500 to 1:180	25,600 – 71,100
Lynch syndrome (*MLH1, MSH2, MSH6, PMS2* and *EPCAM* genes)	1:279	45,900

[a] Adapted from [9-12, 18].
[b] Based on Population Estimates from July 1, 2017, US Census Bureau (Pennsylvania population = 12,805,537).

Guidelines from multiple organizations, including the National Comprehensive Cancer Network (NCCN) [19, 20], the United States Preventive Services Task Force (USPSTF) [21], the CDC Evaluation of Genomic Applications in Practice and Prevention (EGAPP) [22], and joint guidelines from the American College of Medical Genetics and Genomics (ACMG) and the National Society of Genetic Counselors (NSGC) [23] provide recommendations for the identification of individuals at risk for HBOC, LS, and other hereditary cancer syndromes. Health care providers should be educated about these guidelines and encouraged to identify appropriate individuals at risk for HBOC and LS and assure that these at-risk individuals receive accurate and appropriate genetics information, support in the decision-making process, and genetic testing (if appropriate). Providers may wish to consider referring patients to genetic counselors who are healthcare providers with specialized training in medical genetics and counseling skills, for these services.

Genetic information can lead to personalized screening and risk-reduction approaches for individuals with a specific inherited cancer predisposition, as well as targeted (precision) treatment for those with cancer (Figure 1). Individuals identified as having an inherited genetic alteration that results in significantly increased cancer risk (Tables 2 and 3) should be given the opportunity to participate in additional recommended risk management with screening and

risk-reducing interventions and appropriate targeted cancer treatment [19, 20]. These approaches can include, but are not limited to screening at earlier ages, screening at more frequent intervals, utilizing additional screening methods, surgical interventions, and medications.

Figure 1: How Genetics Can Guide Cancer Management

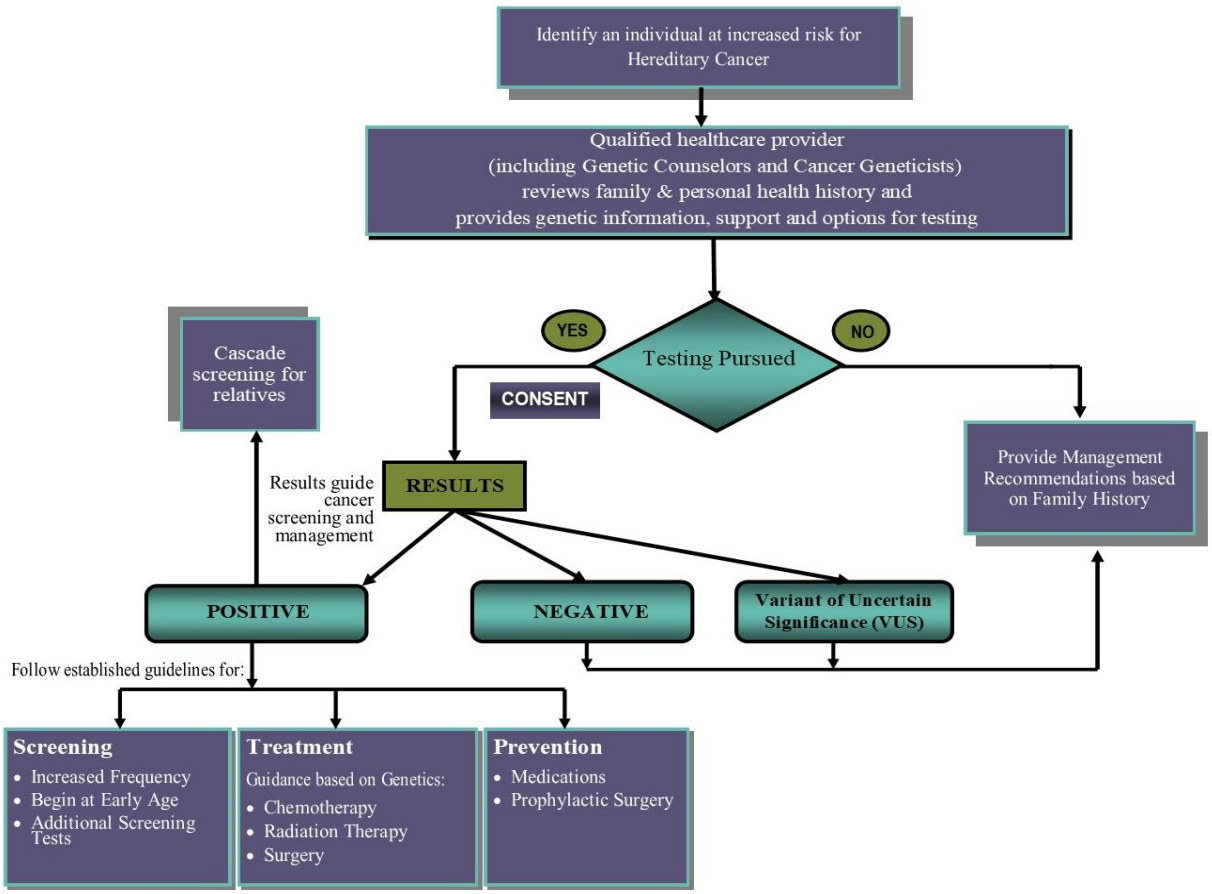


Table 2: Cancer Risks Associated with Hereditary Breast and Ovarian Cancer Syndrome

Hereditary Breast and Ovarian Cancer Syndrome			
Cancer Type	General Population Risk	*BRCA1*	*BRCA2*
Breast	12.3%	46-87%	38-84%
Ovarian	1.3%	39-63%	11-27%
Male Breast	0.1%	1.2%	8.9%
Prostate	6% by 69 11.6% Lifetime	8.6% by 65	20% lifetime risk

Adapted from [12, 24-28]; risk is increased for additional cancers that are not the focus of this report.

Table 3: Cancer Risks Associated with Lynch Syndrome

Lynch Syndrome				
Cancer Type	General Population Risk	*MLH1/MSH2*	*MSH6*	*PMS2*
Colon	4.5%	52-82%	10-22%	15-20%
Endometrial	2.7%	25-60%	16-26%	15%
Ovarian	1.3%	11-24% by age 70	Limited Data	

Adapted from [19, 27]; risk is increased for additional cancers that are not the focus of this report.

Increasingly, genetic information is also being used for therapeutic decision-making. Poly ADP ribose polymerase (PARP) inhibitors have been approved for the treatment of *BRCA1/2*-associated metastatic breast and recurrent ovarian cancers, with additional studies underway [29, 30]. Immune therapy with pembrolizumab is an option for individuals with metastatic cancer in the setting of Lynch syndrome [30]. It is critically important for risk assessment, cancer prevention, and treatment decisions that genetic testing occurs in the appropriate settings.

Utilizing public health strategies to identify and inform individuals who would benefit from genetic counseling and/or testing for hereditary cancer syndromes and then extending testing to relatives of those who test positive for a genetic predisposition via cascade screening provides important opportunities for primary and secondary cancer prevention. As noted earlier, several organizations have published evidence-based guidelines for the identification of individuals with hereditary cancer syndromes [19-23]. However, most individuals with a hereditary cancer syndrome are not identified, potentially amplifying disparities that exist based on race, socioeconomic status, ethnicity, geography, knowledge of genetics, and others [1, 31-34]. Recognizing the unique role of state public health departments in implementing programs to better identify individuals at risk for HBOC and LS, the CDC OPHG has developed a toolkit as a resource to guide state public health efforts [5]. This Tier 1 Genomic Applications Toolkit for Public Health Departments has served as an important resource in the development of the recommendations in this report.

An ad-hoc committee of the CAB was convened to develop recommendations and present them to the CAB in this Report, "Public Health Approach to Cancer Genetics/Genomics in Pennsylvania." The mission of the committee was to investigate and make statewide public health approach recommendations to the Secretary of Health on surveillance to monitor the burden of hereditary cancers, education to address genetic counseling, testing, screening and risk management services, and promotion of policy and systemic change to advance Cancer Genetics/Genomics in the Commonwealth of Pennsylvania to save lives and improve health and quality of life. The committee focused on three main areas for recommendations in the realm of hereditary cancer assessment Data and Surveillance, Genetics Education, and Policy, Insurance Coverage, and Systemic Change.

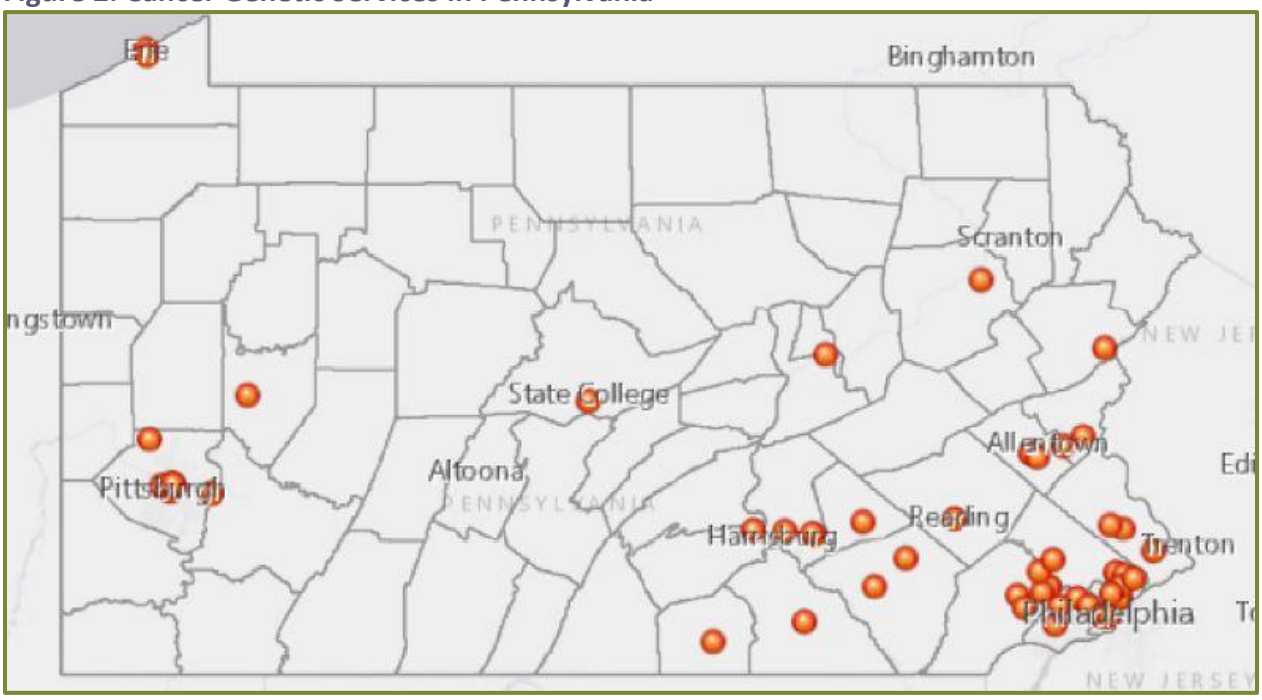
Recommendations

Cancer Genetic Services Data and Surveillance

To determine the progress made by the Pennsylvania Department of Health in improving access to and utilization of cancer genetic services, including genetic counseling, genetic testing, and appropriate screening and prevention methods, assessing the current availability and use of these services as well as robust ongoing surveillance measures are essential. The CDC OPHG has identified surveillance activities as an important component of the "Catalyzing Action" Domain of its Science Impact Framework and has outlined strategies for surveillance for state health departments interested in addressing Tier 1-designated conditions, including HBOC and LS, in its Tier 1 Genomic Applications Toolkit [1, 5]. The Genetics/Genomics Committee focused on these strategies when developing goals relating to Data and Surveillance for Pennsylvania.

The current availability of cancer genetic counseling services in Pennsylvania was determined through an assessment of the American College of Surgeons Commission on Cancer (CoC-accredited hospitals in the state), in addition to data on in-person and telegenetics genetic counseling services from the New York Mid-Atlantic Regional Genetic Network (NYMAC). CoC-accredited hospitals are required to provide genetic services as outlined in Standard 2.3: "Cancer risk assessment, genetic counseling, and genetic testing services are provided to patients either on-site or by referral to a qualified genetics professional" [35]. NYMAC had previously collected genetics clinic information as part of its planned programming for increasing access to genetic services within the region and continues to update this information. A map of cancer genetics providers in Pennsylvania produced by the Cancer Genetics/Genomics committee is shown in Figure 2. The geographic disparities in genetic services evident from the map will be addressed during implementation of the proposed recommendations.

Figure 2: Cancer Genetic Services in Pennsylvania



Multiple states have assessed the public's needs for genetic services through the addition of questions to the state Behavioral Risk Factor Surveillance System (BRFSS) survey specific to cancer genetics/genomics and cancer genetic testing [36]. Adding several cancer genetics questions to the BRFSS would enable assessment across Pennsylvania to understand the needs, and perspectives related to access to genetic counseling and testing services for hereditary cancer risk as well as the surveillance of improvement over time resulting from state-wide programs implemented through the proposed Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health.

The Cancer Genetics/Genomics Committee proposes implementation of the recommendations by coordination between the Cancer Genetics/Genomics Workgroup of the Pennsylvania Cancer Coalition, the Genetics/Genomics Division in the Bureau of Health Promotion and Risk Reduction, of the Pennsylvania Department of Health several additional partners for ongoing data collection regarding inherited cancer syndrome risk and cancer genetics services in Pennsylvania, including the Pennsylvania Cancer Registry, the Pennsylvania Behavioral Risk Factor Surveillance System (BRFSS), the Pennsylvania Medicaid Program, the Pennsylvania Association of Genetic Counselors, and the genetics/genomics clinics and programs in health systems throughout the state, as well as other partners that emerge over time. Additionally, the Division and Workgroup will identify state-specific data in the data sources being used to track the Healthy People 2020 Objectives in Genomics, which include the National Health Interview Survey (NHIS), CDC National Center for Health Statistics, CDC National Center for Chronic Disease Prevention and Health, National Program of Cancer Registries, and the NIH/NCI [37].

Data and Surveillance Recommendations:

The following goals are recommended by the Cancer Genetics/Genomics Committee to address data collection and surveillance related to hereditary cancer syndromes to increase access to and knowledge of genetics and genomics in Pennsylvania among healthcare providers and the public:

1. **Report cancer genetics/genomics data and health disparity data** by:
   a. Enhancing the Behavioral Risk Factor Surveillance System (BRFSS) questionnaire to include several questions about access and uptake of genetic counseling and/or testing, awareness and referral rates.
   b. To incorporate genetic testing information into the state cancer registry.
   c. Assessing available cancer registry data that relate to cancer genetics and genomics to determine future uses of the data (i.e., bi-directional cancer registry reporting).
   d. Establishing state level data from the Healthy People 2020 national data.
   e. Investigating Medicaid claims data related to genetic testing.
2. **Create a Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health** to coordinate implementation of the recommendations of this report, advance the Genetics/Genomics goals of the Cancer Control Plan, and promote awareness of and manage programming to address genetics/genomics issues in Pennsylvania.
3. **Increase access to genetic services in underserved areas in Pennsylvania** by identifying geographic disparities in service, supporting alternate service delivery models for genetics services, identifying funding options to improve access, and pursuing partnerships with State, Regional, and National organizations working towards similar goals (i.e., CDC and the NYMAC Regional Genetics Network).

Education of Healthcare Providers and the Public

Increasing awareness of genetic testing among healthcare providers and the public requires access to appropriate education at several levels. The first step for providers is identifying appropriate candidates for genetic counseling and/or testing. The second step is knowing where to refer these individuals for genetic counseling and testing, if appropriate. Healthcare providers also need support for the aforementioned roles by having access to and obtaining appropriate cancer genetics/genomics training and guidance on long-term follow up for high-risk patients. This committee believes that it is important for the Commonwealth to consolidate, organize, and develop resources to help both patients and providers obtain appropriate cancer genetics/genomics education and guidance at these various levels.

Clinicians must maintain ongoing education in order to deliver safe and effective medical care. Continuing medical and nursing education (CME/CNE) is an essential element of quality care for doctors, nurses, genetic counselors, and other health care providers. The Pennsylvania State Board of Medicine requires 100 credit hours of Continuing Medical Education (CME) every 2 years for physicians [38]. As providers already need to obtain regular education credits, creating CME/CNE-eligible options focusing on cancer genetics topics can be integrated into routine education. Several professional societies, such as the Oncology Nursing Society and the American Society of Clinical Oncology [39] have assembled guidelines for learning objectives in oncology training and continuing education (Table 4).

Table 4. Suggested Genetics/Genomics Learning Objectives for Oncology Education and Continuing Education.[a]

Understanding hereditary predisposition to cancer
Multifactorial nature of cancer risk including genetic and non-genetic risk factors Features unique to hereditary cancer syndromes
Hereditary cancer risk assessment
Review and assessment of family and personal cancer history Pretest consent for cancer susceptibility testing including risks, benefits, and limitations of testing
Implications of genetic testing
Potential genetic testing results, including the possibility of inconclusive results Understanding testing differences between germline vs. somatic tumor profiling Interpretation of incidental and secondary findings from somatic tumor profiling
Review of major hereditary cancer syndromes
Define characteristic tumor spectrum of known syndromes Recognize overlapping phenotypes that generate differential diagnosis for hereditary syndromes based on presenting cancer
Hereditary cancer risk medical management

Apply current recommendations for risk reduction strategies in patients with hereditary cancer syndromes Discuss benefits and limitations of available management strategies Review use of available models for estimating and communicating cancer genetic risks

[a] Adapted from [39].

Table 5. Selected examples of educational resources currently available

Organizations[a]	Website	Description
American Society of Clinical Oncology (ASCO)	www.asco.org/practice-guidelines/cancer-care-initiatives/genetics-toolkit/provider-education-opportunities	10 credit course on cancer genetics and other resources for clinician education
City of Hope	https://www.cityofhope.org/education/health-professional-education/cancer-genetics-education-program/intensive-course-in-cancer-risk-assessment-overview	Intensive course on genetics; great for those practicing in genetics and those without internal resources
National Institutes of Health (NIH)	www.genome.gov/17517037/health-professional-education	Multiple sources of education regarding genetics/genomics
Oncology Nursing Society	www.ons.org/education/courses-activities?combine=genomics	Free and on-demand CNE offerings
American Society of Clinical Oncology (ASCO)	www.asco.org/practice-guidelines/cancer-care-initiatives/genetics-toolkit	Tools and resources to integrate hereditary cancer risk into practice
Cancer Genetics Risk Assessment and Counseling (PDQ®)	www.cancer.gov/about-cancer/causes-prevention/genetics/risk-assessment-pdq#section/_1004	Evidence-based review on topics related to cancer risk assessment
Jackson Laboratories	www.jax.org/education-and-learning/clinical-and-continuing-education/family-history	Family history resources for healthcare providers

[a] Please note: these may not all provide CME/CNE credits.

The development of a regular webinar series would enable ongoing education for providers on the latest updates in cancer genetic testing and high-risk screening. In addition to impacting cancer screening and prevention, genetic testing for hereditary cancer risk has a growing impact on cancer treatment as noted earlier, including surgical, chemotherapy, and

immunotherapy options [40]. Furthermore, somatic tumor testing has become more routine for certain cancer treatment decisions, but the implications for identifying germline hereditary cancer alterations from the results of somatic tumor testing are not always well-understood by providers [41]. Such webinars can be coordinated with thought leaders in the field throughout the state and would provide CME/CNE credit. A certificate of completion for continuing education for non-geneticists could be considered.

Genetic counselors can be identified for both in-person and telegenetics referrals through www.findageneticcounselor.com. However, this resource does not offer a comprehensive collection of appropriately trained genetics providers for cancer risk assessment and long-term follow-up care in the Commonwealth of Pennsylvania. We recommend the development and regular updating of a searchable database of genetics professionals in the Commonwealth, as defined by the Cancer Program Standards on Genetic Counseling and Risk Assessment, for use by both referring healthcare providers and patients.

Patient education and awareness efforts are also important, and we recommend a collection of patient-friendly resources and toolkits within a unified education portal be created. This could include a family history assessment tool and/or a summary of indications within a family history that could suggest consideration of genetic testing. Basic genetics education resources from the Centers for Disease Control (CDC), American Cancer Society (ACS), and other resources can also be included. Patients will also need information to find a genetics provider near them, similar to needs of providers mentioned above, so patients can move forward with genetics consults as appropriate. Public service announcements should be considered to increase awareness of the general public regarding genetic risks and predisposition to common disorders, especially cancer, and available genetic services.

Education Recommendations:

1. **Develop a workgroup in the Pennsylvania Cancer Coalition** to implement recommendations from this report regarding cancer genetics/genomics Education, Data and Surveillance, and Policy, Insurance Coverage and Systemic Change.

2. **Provide ongoing and updated provider education** related to 1) identification and appropriate referrals for at-risk patients, 2) cancer risks related to genetic syndromes, 3) follow up screening/medical management guidelines, and 4) cancer treatment implications associated with hereditary cancer risks. These resources can come in the form of:

   a. A state-supported webpage providing links to education modules, testing guidelines, referral resources, and other relevant topics, e.g., links to National Comprehensive Cancer Network (NCCN), US Preventative Services Task Force (USPSTF), and other guidelines and searchable provider databases, e.g. www.findageneticcounselor.com.

   b. Development of simplified “guides” to assist providers in identifying at-risk patients in their clinics for referrals for genetic counseling/appropriate testing.

   c. Web-based CME/CNE training program, e.g. webinars.

d. In-person training sessions at health centers throughout the state, provided by trained educators and a standardized slide set.

3. **Provide ongoing and updated public education** related to hereditary cancer risk factors and how to find cancer genetics specialists. These resources can come in the form of public service announcements and a State-supported webpage providing links to education modules, testing guidelines, referral resources, e.g., www.findageneticcounselor.com, a statewide provider database, and other relevant topics.

Policy, Insurance and Systemic Change

Development of a Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health would enable synergism within the existing public health infrastructure that maintains the Pennsylvania Cancer Registry, the BRFSS, and carries out cancer control, prevention, research and evaluation. This Division would also work with the CAB and the Genetics/Genomics Workgroup of the Pennsylvania Cancer Coalition to implement the genetics/genomics recommendations in this report and future cancer plans. This would facilitate enhancement of state-wide cancer surveillance activities to address issues involving cancer genetics/genomics. With such infrastructure, the Pennsylvania Department of Health could more readily facilitate education about hereditary cancer risk for the public and healthcare providers. Furthermore, the cancer registry could be leveraged to implement programs meeting Healthy People 2020 genomic objectives to increase the number of individuals who receive genetic counseling and testing for hereditary cancer risk. Identification of individuals with cancer due to hereditary predisposition enables further identification of at-risk family members who have not had cancer, with the goal of prevention or early detection to decrease overall cancer morbidity and mortality in Pennsylvania. Multiple states have created robust genetics/genomics programs within their state public health infrastructure. States such as Michigan, Oregon, Colorado, Utah, Minnesota, Georgia, and Connecticut have programs dedicated to integrating family history tools, surveillance, education, and implementing best practices in cancer genetics/genomics. Through the state public health cancer genetics/genomics infrastructure, these states have also been able to apply for multi-year cooperative agreements with the CDC to expand and improve their cancer-related public health genetics/genomics programs. Policy supporting public health genetics/genomics infrastructure and legislation to fund such infrastructure would enable the Commonwealth to be competitive in securing funds from the CDC and other organizations to improve programs and test innovations needed to close care gaps (such as access in underserved areas), as mentioned above. A new funding opportunity from the CDC Cancer Genomics program has been released recently for Translating Research into Public Health Practice [42].

In addition to implementing and utilizing data from registries and the BRFSS, creation of a public health genetics/genomics program will further facilitate education of state officials, healthcare systems, providers, and the public about genetics/genomics. Leveraging genetics and genomics expertise to educate state officials about issues in public health genetics/genomics and hereditary cancer is needed to improve state laws 1) that currently limit ordering of genetic testing by genetic counselors and 2) protect genomic information. A legal analysis was conducted of all state genetic privacy laws in 2011 [43]. More recently, the laws in three states (Pennsylvania, Ohio and Oregon) were reviewed [44]. Pennsylvania state laws regarding genetic privacy should be reviewed and revised to ensure protections are appropriate for the changing landscape of technology and clinical practice. Pennsylvania had no specific genetic privacy or general health privacy laws in 2011 and has not enacted any since that time. Further review and policymaking are warranted.

Policy, Insurance, and Systemic Change Recommendations:

1. Facilitate legislation to **create a public health genetics/genomics infrastructure** in the Commonwealth of Pennsylvania.
2. Support the proposed **amendment to the genetic counselor licensure law** to allow genetic counselors to order tests.
3. **Seek funding** for a Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health to oversee and manage the implementation of this report, to incorporate genetic testing information into the state cancer registry, and cancer genetic counseling, testing and access to services questions in the BRFSS.

Conclusion

The importance of addressing genetics/genomics as an integral part of a comprehensive plan to decrease cancer incidence, morbidity, and mortality has been established by multiple states and national public health agencies. The Genetics/Genomics Committee views it important that the Commonwealth facilitates the implementation of genetics/genomics goals to enable both patients and providers to obtain appropriate cancer genetics/genomics education and access to genetic services. Inclusion of cancer genetics/genomics goals in the Pennsylvania Cancer Control Plan is an important first step in advancing cancer prevention, early detection, and treatment efforts for individuals at the highest risk for cancer in the state. However, the Genetics/Genomics Committee recognizes that implementing programs and activities related to these goals and ultimately increasing the use of and access to genetic services requires a long-term commitment from the Commonwealth as well as coordination between experts in genetics, genomics, oncology, and public health across Pennsylvania.

The Genetics/Genomics Committee recommends that implementation of the recommendations outlined in this report begin by establishing a permanent Genetics/Genomics Workgroup in the Pennsylvania Cancer Coalition. Establishing this workgroup will initiate the coordination of efforts from genetics providers, public health professionals, advocacy organizations, and others in Pennsylvania working to advance these goals. As part of the Pennsylvania Cancer Coalition, the Genetics/Genomics Workgroup will have the opportunity to collaborate with other workgroups and committees to further the goals outlined in the Cancer Control Plan in genetics/genomics and across additional specialties.

While goals and projects are expected to evolve over time, this committee, in conjunction with additional stakeholders working as part of the Genetics/Genomics Workgroup, is prepared to begin work on the following:

- Investigating available data sources and partnerships for expanding the assessment of the burden of hereditary cancers for individuals, families and communities, and the utilization of genetic counseling, genetic testing and associated clinical services in Pennsylvania.
- Developing and organizing multimedia educational tools for the public and health care providers about hereditary cancer and genetic counseling, genetic testing and associated clinical services in Pennsylvania.
- Identifying and working with partners in insurance, public policy, and other areas to begin discussions about initiatives to improve access to and insurance coverage of genetic services for individuals and families at increased risk for hereditary cancer syndromes.

In order to coordinate the efforts of multiple stakeholders and further advance the integration of genetics and genomics into public health initiatives in the Commonwealth, the Genetics/Genomics Committee has recommended establishing a Genetics/Genomics Division within the Bureau of Health Promotion and Risk Reduction of the Pennsylvania Department of Health. As outlined in this report, the successful utilization of cancer genetics and genomics to influence primary, secondary and tertiary prevention of cancer requires coordinated efforts in data/surveillance, education, and policy/insurance in addition to other factors not addressed

due to the scope of this report. Implementation of programming and competitive application for funding will require the coordination of multiple Department of Health Bureaus and Divisions in the existing Pennsylvania public health infrastructure as well as with genetics providers, advocacy and professional organizations, parent and family organizations, universities, health care systems, and insurance companies in Pennsylvania. Furthermore, ongoing collaboration with regional and national organizations, agencies, and initiatives working to advance public health genetics/genomics goals will be essential to synergizing the work in Pennsylvania with these larger efforts. The Cancer Genetics/Genomics committee believes that this will best be accomplished through a permanent Department of Health Genetics/Genomics Division able to address the significance of genetics and genomics in cancer control as well as other health conditions and public health initiatives in Pennsylvania. Creating this Division will also help facilitate the ongoing education of stakeholders and the public in Pennsylvania concerning new information about genetics/genomics as well as to coordinate policy development in this area.

The "Public Health Approach to Cancer Genetics/Genomics in Pennsylvania" report, developed by the CAB Cancer Genetics/Genomics Committee outlines a preliminary assessment of the burden of hereditary cancer and availability of genetic services in Pennsylvania as well as proposed recommendations focusing on data/surveillance, education, and policy, insurance and systemic change to address hereditary cancer in Pennsylvanians. We recommend that these recommendations form the basis of the Genetics/Genomics Goals in the *2019-2023 Pennsylvania Cancer Control Plan*. The Genetics/Genomics committee is pleased to provide this report to the CAB for review and approval prior to submission to the Pennsylvania Department of Health leadership and associated state agencies to help us reach our patients/families in need of this care and to best serve our communities.

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