

Cancer Genetics in Connecticut: How Does It Fit into the Comprehensive Cancer Plan and Where Do We Go from Here?

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Since the publication of the *Connecticut Cancer Plan, 2014-2017* genetic risk assessment has become a significant component of the cancer control process. The work of the Connecticut Department of Public Health Genomics Office (DPH-GO) focuses on recommendations related to cancer genetics. The 2013 US Preventive Services Task Force¹ (USPSTF) (mentioned on p. 37 of the Plan) recommends that Primary Care Providers (PCPs) screen all women for family history of conditions associated with an increased risk for potential harmful mutations in the breast/ovarian cancer genes *BRCA1* and *BRCA2*, and that those women with a positive family health history be referred for genetic counseling and then, if indicated, for genetic testing. The 2009 Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group² recommendations concerning Lynch syndrome are that genetic testing be offered to persons newly diagnosed with colorectal cancer to reduce morbidity and mortality in their relatives. The DPH-GO works with providers around the state to encourage implementation of the USPSTF and EGAPP Guidelines, and to assure that high quality, equitable genetics services are available to all Connecticut residents.

Example Family History Screening Tool

Did any of your first-degree relatives have breast or ovarian cancer?

Did any of your relatives have bilateral breast cancer?

Did any man in your family have breast cancer?

Did any woman in your family have breast and ovarian cancer?

Did any woman in your family have breast cancer before age 50?

Do you have two or more relatives with breast and/or ovarian cancer?

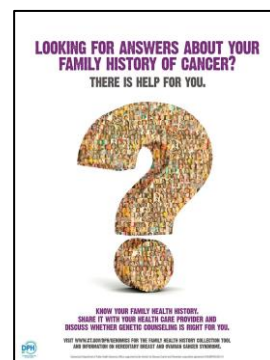
Do you have two or more relatives with breast and/or bowel cancer?

Identification of individuals and families at high risk for breast, ovarian and colorectal cancer, and referral to genetic counseling and testing fit under Goal #2 of the Plan: "High quality cancer screening and early detection services are available to all people living in Connecticut." Activities of the DPH-GO address both objectives under this goal: (1) Support policy, systems and environmental changes that increase the percentage of Connecticut residents receiving recommended and appropriate breast, cervical, colorectal and lung screening; and (2) Identify and disseminate strategies and best practices to reduce disparities in access to screening services.

Although not explicitly stated in the discussion of the Affordable Care Act³ (p. 24 in the Plan) genetic services when appropriate are covered under the following provision: "Coverage is guaranteed and out-of-pocket costs are eliminated in new insurance plans for proven preventive and screening services, giving people access to lifesaving screenings for breast, cervical and colorectal cancer." The latest interpretation of this mandate is that new insurance plans are required to cover both genetic counseling and testing, when appropriate, based on family history.⁴

Goal #3 of the Plan states "High quality comprehensive cancer treatment and the opportunity to participate in clinical trials are available and accessible to all people living in Connecticut." As part of this goal, Objective #3.1 is to "[p]romote and support the efforts of Connecticut hospitals to meet the standards of the American College of Surgeons' Commission on Cancer (CoC) Standard 2.3⁵ (p. 39 in the Plan) is relevant - "Cancer risk assessment, genetic counseling and testing services are provided to patients either on-site or by referral to a qualified genetics professional." One of the "Strategic Actions" identified under this Goal is to "[s]upport efforts to make information on underlying genetic/heritable causes of common cancers and the importance of genetic counseling and early detection more readily available to the public." DPH-GO is focused on incorporating evidence-based cancer genomics best practices into primary care through public health practice.

PCPs are expected to play an increasing role in the identification and referral of patients at high risk for hereditary cancers. A major initiative, funded by the Centers for Disease Control and Prevention's Division of Cancer Prevention and Control, addresses this issue via several avenues. For example, earlier this year, the document [Cancer Genomics Best Practices for Connecticut Healthcare Providers](#) was updated and posted on the DPH website. A poster (see inset) about cancer genetic counseling and testing, for hanging in the



The Connecticut Cancer Partnership is the coalition recognized by the US Centers for Disease Control and Prevention (CDC) to implement the concepts of comprehensive cancer control through collaborative planning and prioritized activities with its member organizations. It has developed a series of multi-year state cancer plans. The Partnership has membership representing over 150 organizations in Connecticut with cancer control as their common interest.

offices of PCPs and others, has been created and distributed. The “Mentoring Cancer Genomics in Primary Care” project (October 2015 – August 2016) aims to educate providers in this rapidly evolving field and to encourage discussion of real-life clinical cases with the goal of improving quality and quantity of referrals to genetic service providers. This program places genetic counselors with cancer expertise in the offices of PCPs around the state to provide in-person training, using a curriculum developed for this project consisting of a series of nine modules (see inset) on topics such as “Hereditary Breast and Ovarian Cancer Risk Factors and National Guidelines”, “Tools to Support HBOC Risk Assessment” (see example tool in box) and “Implications for Relatives – Cascade Screening and the Role of the PCP”.

The curriculum has been piloted; suggestions from providers participating in evaluation of the modules will be incorporated into the next iteration of the slide set and the series implemented with a new set of providers in the future. Because women and men of Ashkenazi (Eastern European) Jewish ethnicity are at higher risk to carry a BRCA mutation, the DPH-GO sponsored a panel discussion at the University of Connecticut Hillel House on November 4, 2015 to inform students about risks and preventive options and to encourage them to share this information with their relatives. Finally, the Genomics Office is working with Connecticut’s Breast and Cervical Cancer Early Detection Program staff to incorporate questions on personal and family history of cancer into their intake forms to assure that high-risk women served through this program reap the benefits associated with genetic counseling and testing. As a first step, key informants at the 11 Connecticut sites providing such services were interviewed to learn about current practices.

The DPH-GO is committed to creating new partnerships and to expanding our programs to new areas of the state. Those interested in learning more should visit the DPH-GO website or e-mail the staff at DPH.Genomics@ct.gov.

Curriculum Modules

Introduction

HBOC Risk Factors and National Guidelines

Role of the PCP

Tools to Support HBOC Risk Assessment

Genetic Counseling and Testing

Management of Individuals with BRCA1 or 2 Mutations or VUS

Implications for Relatives – Cascade Screening

Ethical, Legal and Financial Issues

Future Directions

References:

- ¹ U.S. Preventive Services Task Force. Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer in Women: U.S. Preventive Services Task Force Recommendation Statement. *Ann Intern Med.* 2014; 160:271-281.
- ² Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group. 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.* 2009; 11:35-41.
- ³ Patient Protection and Affordable Care Act. 42 U.S.C. §18001 et seq. (2010)
- ⁴ Affordable Care Act Implementation FAQs – Set 12. The Center for Consumer Information and Insurance Oversight, Centers for Medicare and Medicaid Services. https://www.cms.gov/CCIIO/Resources/Fact-Sheets-and-FAQs/aca_implementation_faqs12.html
- ⁵ American College of Surgeons Commission on Cancer. 2012. Cancer Program Standards 2012: Ensuring Patient-Centered Care, v1.2.1. Pages 68-69: Standard 2.3 Risk Assessment and Genetic Counseling. Chicago: American College of Surgeons. <https://www.facs.org/~media/files/quality%20programs/cancer/coc/programstandards2012.ashx>