

# CANCER GENOMICS BEST PRACTICES

*for Connecticut Healthcare Providers*



**Hereditary  
Breast and Ovarian  
Cancer Syndrome**

**Lynch Syndrome**



CONNECTICUT DEPARTMENT OF PUBLIC HEALTH  
2015

STATE OF CONNECTICUT  
DEPARTMENT OF PUBLIC HEALTH

Jewel Mullen, M.D., M.P.H., M.P.A.  
Commissioner



Dannel P. Malloy  
Governor  
Nancy Wyman  
Lt. Governor

December 16, 2015

Dear Healthcare Partner:

The Connecticut Department of Public Health is providing information to healthcare providers and each acute care hospital in the state about two hereditary syndromes that predispose individuals to cancer. They are hereditary breast and ovarian cancer (HBOC) syndrome and Lynch syndrome (formerly called Hereditary Non-Polyposis Colorectal Cancer Syndrome).

The following information booklet, *Cancer Genomics Best Practices for Connecticut Healthcare Providers*, contains:

- A statewide report from the DPH Genomics Office and Connecticut Tumor Registry on cancers diagnosed in patients who may be at increased risk for HBOC or Lynch syndromes.
- Evidence-based guidelines related to HBOC (due to *BRCA* mutations) and Lynch syndrome.
- A list of cancer genetic counselors in Connecticut.
- Information about genetic and family health history resources, including a Patient Information sheet for the two syndromes.

These materials are made possible by a grant to DPH from the Centers for Disease Control and Prevention (CDC), Cooperative Agreement #DP5355-01. The purpose is to inform Connecticut health care practitioners about evidence-based recommendations for referral to, and use of, cancer genomic services.

Please use this information to assist in your efforts to improve health outcomes for your patients and their families. As DPH moves forward with its state health improvement planning process, we will continue to promote similar evidence-based practices focused on prevention.

Beverly Burke, Genomics Office Coordinator is available to discuss these materials. Ava Nepaul, Epidemiologist, is available to discuss the data reports. You may contact them at [DPH.Genomics@ct.gov](mailto:DPH.Genomics@ct.gov).

Thank you for helping to promote cancer genomics best practices in our state.

Sincerely,

A handwritten signature in black ink that reads "Jewel Mullen".

Jewel Mullen, MD, MPH, MPA  
Commissioner

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# CANCER GENOMICS BEST PRACTICES

## for Connecticut Healthcare Providers

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*The Public Health Genomics Office staff gratefully acknowledge the following persons who contributed data, technical assistance, and/or critical review of the text.*

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Thanks to the members of the Expert External Genomics Advisory Panel and DPH Council on Genomics who support and inform the work of the Public Health Genomics Office.

Permission to copy, distribute, or otherwise use information from this publication is hereby granted. Appropriate acknowledgement of the source is requested, using the following citation:  
Connecticut Department of Public Health Genomics Office. 2015. *Cancer Genomics Best Practices for Connecticut Healthcare Providers: Hereditary Breast and Ovarian Cancer Syndrome and Lynch Syndrome*. Hartford, CT: Connecticut Department of Public Health.

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The Connecticut Department of Public Health encourages the adoption by healthcare practitioners of national, evidence-based guidelines for genetic counseling and testing concerning *BRCA*-related hereditary breast and ovarian cancer (HBOC) and Lynch syndromes. Such recommendations reflect increasing scientific evidence supporting the health benefits of using family health history and genetic testing to guide clinical assessments and enable implementation of preventive measures.

The present initiative is consistent with the U.S. Department of Health and Human Services Healthy People 2020 Genomics objectives and evidence-based guidelines/recommendations from: the U.S. Preventive Service Task Force (USPSTF); National Comprehensive Cancer Network® (NCCN®); and Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group.

### HEALTHY PEOPLE 2020 GENOMICS OBJECTIVES

**Objective G-1:** Increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling.

**Objective G-2:** (Developmental) Increase the proportion of persons with newly diagnosed colorectal cancer who receive genetic testing to identify Lynch syndrome (or familial colorectal cancer syndromes).

#### HBOC SYNDROME

##### USPSTF Recommendation Statement, 2013<sup>a</sup>

*The USPSTF recommends that primary care providers screen women who have family members with breast, ovarian, tubal, or peritoneal cancer with one of several screening tools designed to identify a family history that may be associated with an increased risk for potential harmful mutations in breast cancer susceptibility genes (BRCA1 and BRCA2). Women who screen positive should receive genetic counseling and, if indicated after counseling, BRCA testing.*

*The USPSTF recommends against routine genetic counseling or BRCA testing of women whose family history is not associated with an increased risk for potentially harmful mutations in the BRCA1 or BRCA2 genes.*

<sup>a</sup>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.

#### LYNCH SYNDROME

##### EGAPP Working Group Recommendation Statement, 2009<sup>b</sup>

*The EGAPP Working Group (EWG) found sufficient evidence to recommend offering genetic testing for Lynch syndrome to individuals with newly diagnosed colorectal cancer to reduce morbidity and mortality in relatives.*

*We [EWG] found insufficient evidence to recommend a specific genetic testing strategy among the several examined.*

<sup>b</sup> 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.

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Published November 2015

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## CONTENTS

<b>Genetics: Important terms</b>	<b>1</b>
<b>1 Hereditary Breast and Ovarian Cancer Syndrome (HBOC Syndrome)</b>	<b>2</b>
▪ Report: Potential Cases of HBOC Syndrome, Connecticut, 2010 – 2011	2
▪ Information for Connecticut Healthcare Professionals: <i>HBOC Syndrome</i>	3
▪ Summary of Evidence-based Clinical Practice Guidelines: <i>Genetic Susceptibility Testing for HBOC Syndrome</i>	
◆ U.S. Preventive Services Task Force	5
◆ National Comprehensive Cancer Network®	6
▪ Information for Patients and Families: <i>HBOC Syndrome</i>	9
<b>2 Lynch Syndrome (LS) or Hereditary Nonpolyposis Colorectal Cancer (HNPCC)</b>	<b>12</b>
▪ Report: <i>Potential cases of Lynch Syndrome, Connecticut, 2010 – 2011</i>	12
▪ Information for Connecticut Healthcare Professionals: <i>Lynch Syndrome</i>	13
▪ Summary of Evidence-based Clinical Practice Guidelines: <i>Genetic Susceptibility Testing for Lynch Syndrome</i>	15
◆ Evaluation of Genomic Applications in Practice and Prevention Working Group	
▪ Information for Patients and Families: <i>Lynch Syndrome</i>	17
<b>3 Resources for Health Professionals and Consumers</b>	<b>19</b>
▪ American College of Surgeons Commission on Cancer: <i>Standard 2.3 – Risk Assessment and Genetic Counseling</i>	19
▪ Provider Action Plan	21
▪ Cancer Genetic Counselors in Connecticut	22
▪ Additional Resources	24
▪ References	27
▪ Connecticut Family Health History Pocket Guide	29

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# Genetics: Important terms

This list of terms is provided so that healthcare professionals and members of the general public who use this resource can quickly access the definitions of terms and concepts that appear in this publication. The definitions are adapted from the National Library of Medicine's Genetics Home Reference (<http://ghr.nlm.nih.gov/>).

TERM/CONCEPT	DEFINITION
Autosomal dominance	A pattern of inheritance characteristic of some genetic conditions (e.g., Hereditary Breast and Ovarian Cancer syndrome, Lynch syndrome). "Autosomal" means that the gene in question is located on one of the numbered, or non-sex, chromosomes. "Dominant" means that a single copy of the disease-associated mutation is enough to cause the disease. This is in contrast to a recessive disorder, where two copies of the mutation are needed to cause the disease.
DNA	Deoxyribonucleic acid. The hereditary materials in humans and almost all other organisms. It is composed of four chemical bases. The order, or sequence, of the bases determines the information available for building and maintaining an organism, similar to the way in which letters of the alphabet appear in a certain order to form words and sentences.
Gene	The basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to make molecules called proteins. Every person has two copies of each gene, one inherited from each parent.
Genetics	The branch of biology that deals with heredity, especially the mechanisms of hereditary transmission and the variation of inherited characteristics among similar or related organisms.
Genetic predisposition	An increased likelihood of developing a particular disease based on a person's genetic makeup (sometimes also called genetic susceptibility). A genetic predisposition results from specific genetic variations that are often inherited from a parent. These genetic changes contribute to the development of a disease but do not directly cause it. Some people with a predisposing genetic variation will never get the disease while others will, even within the same family.
Mutation	A mutation is a change in a DNA sequence. Mutations can result from DNA copying mistakes made during cell division, exposure to ionizing radiation, exposure to chemicals called mutagens, or infection by viruses. Germ line mutations occur in the eggs and sperm and can be passed on to offspring, while somatic mutations occur in body cells and are not passed on.

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# Potential Cases of Hereditary Breast and Ovarian Cancer Syndrome Connecticut, 2010-2011

Cancer site	Number of cancer patients <sup>a</sup>
Female breast ( $\leq$ 50 yrs. of age)	1,427
Ovary (All ages)	576
Male breast	63
Multiple primary sites <sup>b</sup> (Breast-breast or breast-ovary)	629
Triple negative breast cancer <sup>c</sup>	612

Source: Connecticut Tumor Registry.

<sup>a</sup> Patients who were Connecticut residents at the time of diagnosis. Invasive cancers only. Includes death certificate-only registrations.

<sup>b</sup> More than one cancer diagnosis, with the most recent diagnosis in 2010-2011.

<sup>c</sup> According to the National Cancer Institute, this term “describes breast cancer cells that do not have estrogen receptors, progesterone receptors, or large amounts of HER2/neu protein. Also called ER-negative PR-negative HER2/neu-negative breast cancer.”

Please note: Data in this table are not comparable to the data published in the 2012 version of this document. This table shows the number of **patients** and not the number of tumors.

**This table contains the number of patients diagnosed during 2010 – 2011 who may be more likely to have a breast or ovarian cancer-causing mutation such as *BRAC1* or *BRAC2*.**

## WHY THESE DATA ARE IMPORTANT FOR YOU TO KNOW

Hereditary Breast and Ovarian Cancer syndrome (HBOC) is a collective term that describes genetic susceptibility to breast and/or ovarian cancer. HBOC accounts for approximately 5-10% of all breast cancer diagnoses and 10-15% of all ovarian cancer diagnoses. Currently, mutations in the tumor suppressor genes *BRAC1* and *BRCA2* are the most commonly identified causes of HBOC.

### A woman with HBOC syndrome has an increased lifetime risk of certain cancers.

- Up to 80% risk of breast cancer (compared to 12-13% in the general population).
- Up to 40% risk of ovarian cancer (compared to 1-2% in the general population).
- A 40% risk of a second primary breast cancer within 10 years of first diagnosis.

### Any of the following diagnoses increase the likelihood of HBOC due to a *BRCA1* or *BRCA2* mutation.

- Early onset female breast cancer (diagnosed at or before 50 years of age).
- Ovarian cancer (diagnosed at any age).
- Male breast cancer.
- Two or more of any combination of breast and/or ovarian cancers in the same patient.

### Patients with any of the above diagnoses should be considered for a formal risk assessment and genetic counseling and/or testing.

- Refer patients to a health care provider who is suitably trained in cancer genetics.
- Individuals with breast or ovarian cancer who are diagnosed with HBOC may have a higher risk for developing future cancers and may benefit from closer monitoring or special medical management.
- Biological relatives of these individuals also may be affected.

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## Hereditary Breast and Ovarian Cancer Syndrome

### What is Hereditary Breast and Ovarian Cancer (HBOC) syndrome?

HBOC syndrome is a hereditary cancer predisposition condition usually caused by harmful mutations in the *BRCA1* and *BRCA2* genes (breast cancer susceptibility genes 1 and 2). *BRCA1* and *BRCA2* belong to a class of genes known as tumor suppressors. In normal cells, *BRCA1* and *BRCA2* help ensure the stability of the cell's genetic material (DNA) and help prevent uncontrolled cell growth. A mutated *BRCA* gene can be inherited from either parent.

### Facts about HBOC syndrome

- Approximately 5% to 10% of breast cancer patients have HBOC syndrome.
- Approximately 10% to 15% of ovarian cancer patients have HBOC syndrome.
- Women who inherit an altered *BRCA1* and/or *BRCA2* gene have a 40% to 80% lifetime risk of being diagnosed with breast cancer, compared to 12% to 13% risk for women in the general population.
- Women who inherit an altered *BRCA1* and/or *BRCA2* gene have a 11% to 40% lifetime risk of being diagnosed with ovarian cancer, compared to a 1% to 2% risk for women in the general population. Although ovarian cancer is less common than breast cancer, it is more often fatal.
- Women with a harmful *BRCA* genetic variant are more likely than non-carriers to be diagnosed with cancer before 50 years of age.
- Men with a *BRCA1* and/or *BRCA2* mutation have a 5% to 10% risk of being diagnosed with breast cancer, compared to a 0.1% risk for men in the general population.
- The lifetime risk for prostate cancer in men with altered *BRCA1* or *BRCA2* genes is 30% to 39%.

### How common are *BRCA1* and *BRCA2* mutations?

- An estimated 1 in 300 to 1 in 800 individuals in the general population carries a *BRCA1* or *BRCA2* mutation.
- Among individuals of Ashkenazi (Eastern European) Jewish ancestry, 1 in 40 individuals carries a *BRCA1* or *BRCA2* mutation.

### Who is most likely to have a *BRCA1* or *BRCA2* mutation?

- The likelihood is highest in families with: a history of multiple cases of breast cancer; cases of both breast and ovarian cancer; one or more family members with two primary cancers (original tumors that develop at different sites in the body); or Ashkenazi Jewish ancestry.
- Not every woman or man in such families, however, carries a harmful *BRCA1* or *BRCA2* mutation, and not every cancer in such families is linked to a harmful mutation in one of these genes; rather, the cancers may result from sporadic somatic mutations. Furthermore, not every person who has a harmful *BRCA1* or *BRCA2* mutation will develop breast and/or ovarian cancer.

### Why is genetic counseling important?

Genetic counseling helps people better understand their risk for hereditary cancer, so they can make informed decisions about genetic testing and follow-up care.

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## Hereditary Breast and Ovarian Cancer Syndrome

### What does genetic counseling involve?

Genetic counseling encompasses the following services:

- Reviewing an individual’s personal history and detailed family medical history.
- Assessing and explaining risk for hereditary cancers and the chance of finding a mutation through genetic testing.
- Discussing the benefits, limitations, and other possible consequences of genetic testing.
- Outlining medical implications of a positive or a negative test result.
- Determining which biological relative is most appropriate to begin the genetic testing process.
- Interpreting genetic test results, and explaining what they mean for individuals and their relatives.
- Providing referrals to experts for follow-up screening and risk management.
- Providing referrals to support resources and research opportunities (including research on genetic testing, screening, treatment, etc.).
- Discussing risks and medical management options with a patient’s other health care provider(s).
- Addressing common concerns about the privacy and confidentiality of personal genetic information.

### What are the benefits of genetic testing for HBOC syndrome?

- A positive test result can bring relief from uncertainty. It helps people to make informed decisions about their futures, and allows them to take steps to reduce their cancer risk through increased surveillance or other medical and lifestyle choices.
- A positive test result may help to explain why individuals or biological relatives had cancer in the past, and, should they choose to share test results, may provide their family members with useful information.
- Those who have a positive test result may be able to participate in medical research that could, in the long run, help reduce deaths from breast, ovarian, and prostate cancer.
- A negative test result may provide a sense of relief and preclude the need for additional procedures or tests beyond routine cancer screening.

### What are the disadvantages of genetic testing for HBOC syndrome?

Test results may affect a person’s emotions, family relationships, finances, privacy, and medical choices.

- A positive result may make a person feel anxious, angry, or depressed. Medical treatments, such as surgery to try to prevent the cancer, could have serious, long-term implications and uncertain effectiveness.
- A negative result may make people feel guilty about escaping a disease that affected one or more loved ones (“survivor guilt”).
- People may get a false sense of security that they have no chance of getting cancer, when their cancer risk is likely the same as that of the general population.
- Because genetic testing can reveal information about more than one family member, the emotions caused by the results can create tension within families. The results also can affect personal choices, such as marriage and childbearing.
- Privacy and confidentiality of genetic test results are additional potential concerns. HIPAA and GINA are federal acts which address the protection of genetic information.
- Genetic testing can be expensive depending on the extent of testing. Many insurance plans cover the cost of the testing for persons at high risk.

**NOTE:** Specific indicators for genetic counseling and testing vary among professional organizations. Guidelines are not a substitute for clinical judgment. Not all clinical scenarios can be anticipated, such as when there is limited family structure or incomplete family medical history.

# Summary of Evidence-based Clinical Practice Guidelines: Genetic Susceptibility Testing for Hereditary Breast and Ovarian Cancer Syndrome

## USPSTF

- **Women who have family members with breast, ovarian, tubal, or peritoneal cancer**
  - ◆ Screen women whose family history may be associated with an increased risk for potentially harmful *BRCA* mutations.
  - ◆ Women with positive screening results should receive genetic counseling and, if indicated after counseling, *BRCA* testing.
- **Women whose family history is not associated with an increased risk for potentially harmful *BRCA* mutations**
  - ◆ Do not routinely recommend genetic counseling or *BRCA* testing.

Source: 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(4):271-281. For additional information, see [www.uspreventiveservicestaskforce.org](http://www.uspreventiveservicestaskforce.org)



### RISK ASSESSMENT, GENETIC COUNSELING, AND GENETIC TESTING FOR *BRCA*-RELATED CANCER IN WOMEN CLINICAL SUMMARY OF U.S. PREVENTIVE SERVICES TASK FORCE RECOMMENDATION

Population	Asymptomatic women who have not been diagnosed with <i>BRCA</i> -related cancer	
Recommendation	<p>Screen women whose family history may be associated with an increased risk for potentially harmful <i>BRCA</i> mutations. Women with positive screening results should receive genetic counseling and, if indicated after counseling, <i>BRCA</i> testing.</p> <p>Grade: B</p>	<p>Do not routinely recommend genetic counseling or <i>BRCA</i> testing to women whose family history is not associated with an increased risk for potentially harmful <i>BRCA</i> mutations.</p> <p>Grade: D</p>
Risk Assessment	<p>Family history factors associated with increased likelihood of potentially harmful <i>BRCA</i> mutations include breast cancer diagnosis before age 50 years, bilateral breast cancer, family history of breast and ovarian cancer, presence of breast cancer in <math>\geq 1</math> male family member, multiple cases of breast cancer in the family, <math>\geq 1</math> or more family member with 2 primary types of <i>BRCA</i>-related cancer, and Ashkenazi Jewish ethnicity.</p> <p>Several familial risk stratification tools are available to determine the need for in-depth genetic counseling, such as the Ontario Family History Assessment Tool, Manchester Scoring System, Referral Screening Tool, Pedigree Assessment Tool, and FHS-7.</p>	
Screening Tests	<p>Genetic risk assessment and <i>BRCA</i> mutation testing are generally multistep processes involving identification of women who may be at increased risk for potentially harmful mutations, followed by genetic counseling by suitably trained health care providers and genetic testing of selected high-risk women when indicated.</p> <p>Tests for <i>BRCA</i> mutations are highly sensitive and specific for known mutations, but interpretation of results is complex and generally requires posttest counseling.</p>	
Treatment	<p>Interventions in women who are <i>BRCA</i> mutation carriers include earlier, more frequent, or intensive cancer screening; risk-reducing medications (e.g., tamoxifen or raloxifene); and risk-reducing surgery (e.g., mastectomy or salpingo-oophorectomy).</p>	
Balance of Benefits and Harms	<p>In women whose family history is associated with an increased risk for potentially harmful <i>BRCA</i> mutations, the net benefit of genetic testing and early intervention is moderate.</p>	<p>In women whose family history is not associated with an increased risk for potentially harmful <i>BRCA</i> mutations, the net benefit of genetic testing and early intervention ranges from minimal to potentially harmful.</p>
Other Relevant USPSTF Recommendations	<p>The USPSTF has made recommendations on medications for the reduction of breast cancer risk and screening for ovarian cancer. These recommendations are available at <a href="http://www.uspreventiveservicestaskforce.org">www.uspreventiveservicestaskforce.org</a>.</p>	

For a summary of the evidence systematically reviewed in making this recommendation, the full recommendation statement, and supporting documents, please go to [www.uspreventiveservicestaskforce.org](http://www.uspreventiveservicestaskforce.org).

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# Summary of Evidence-based Clinical Practice Guidelines:

## Genetic Susceptibility Testing for Hereditary Breast and Ovarian Cancer Syndrome

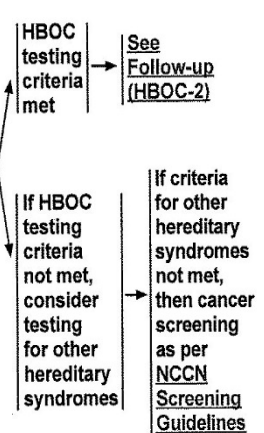
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#### HEREDITARY BREAST AND/OR OVARIAN CANCER SYNDROME TESTING CRITERIA<sup>a,b</sup>

Meeting one or more of these criteria warrants further personalized risk assessment, genetic counseling, and often genetic testing and management. Testing of unaffected individuals should only be considered when an appropriate affected family member is unavailable for testing.

- Individual from a family with a known deleterious *BRCA1/BRCA2* mutation
- Personal history of breast cancer<sup>b</sup> + one or more of the following:
  - ▶ Diagnosed ≤45 y
  - ▶ Diagnosed ≤50 y with:
    - ◊ An additional breast cancer primary<sup>c</sup>
    - ◊ ≥1 close blood relative<sup>d</sup> with breast cancer at any age
    - ◊ ≥1 close relative with pancreatic cancer
    - ◊ ≥1 relative with prostate cancer (Gleason score ≥7)
    - ◊ An unknown or limited family history<sup>a</sup>
  - ▶ Diagnosed ≤60 y with a:
    - ◊ Triple negative breast cancer
  - ▶ Diagnosed at any age with:
    - ◊ ≥1 close blood relative<sup>d</sup> with breast cancer diagnosed ≤50 y
    - ◊ ≥2 close blood relatives<sup>d</sup> with breast cancer at any age
    - ◊ ≥1 close blood relative<sup>d</sup> with invasive ovarian<sup>e</sup> cancer
    - ◊ ≥2 close blood relatives<sup>d</sup> with pancreatic cancer and/or prostate cancer (Gleason score ≥7) at any age
    - ◊ A close male blood relative<sup>d</sup> with breast cancer
    - ◊ For an individual of ethnicity associated with higher mutation frequency (eg, Ashkenazi Jewish) no additional family history may be required<sup>f</sup>
- Personal history of invasive ovarian<sup>e</sup> cancer
- Personal history of male breast cancer
- Personal history of prostate cancer (Gleason score ≥7) at any age with ≥1 close blood relative<sup>d</sup> with breast (≤50 y) and/or invasive ovarian<sup>e</sup> and/or pancreatic or prostate cancer (Gleason score ≥7) at any age
- Personal history of pancreatic cancer at any age with ≥1 close blood relative<sup>d</sup> with breast (≤50 y) and/or invasive ovarian<sup>e</sup> and/or pancreatic cancer at any age
- Personal history of pancreatic cancer, and Ashkenazi Jewish ancestry
- Family history only (significant limitations of interpreting test results for an unaffected individual should be discussed):
  - ▶ First- or second-degree blood<sup>d</sup> relative meeting any of the above criteria
  - ▶ Third-degree blood<sup>d</sup> relative who has breast cancer<sup>b</sup> and/or invasive ovarian<sup>e</sup> cancer and who has ≥2 close blood relatives<sup>d</sup> with breast cancer (at least one with breast cancer ≤50 y) and/or invasive ovarian<sup>f</sup> cancer



<sup>a</sup>For further details regarding the nuances of genetic counseling and testing, see [BR/OV-A](#).

<sup>b</sup>For the purposes of these guidelines, invasive and ductal carcinoma in situ breast cancers should be included.

<sup>c</sup>Two breast cancer primaries includes bilateral (contralateral) disease or two or more clearly separate ipsilateral primary tumors either synchronously or asynchronously.

<sup>d</sup>Close blood relatives include first-, second-, and third-degree relatives on same side of family. (See [BR/OV-B](#))

<sup>e</sup>Includes fallopian tube and primary peritoneal cancers. *BRCA*-related ovarian cancers are associated with epithelial non-mucinous histology. Other cancer genetic syndromes may be associated with mucinous ovarian cancer. Non-epithelial ovarian cancer may be associated with PJS and possibly other cancer syndromes. Ovarian/fallopian tube/primary peritoneal cancers are component tumors of Lynch syndrome; be attentive for clinical evidence of this syndrome. See [NCCN Guidelines for Genetic/Familial High-Risk Assessment: Colorectal](#).

<sup>f</sup>Testing for Ashkenazi Jewish founder-specific mutation(s) should be performed first. Comprehensive genetic testing may be considered if ancestry also includes non-Ashkenazi Jewish relatives or if other HBOC criteria are met. Founder mutations exist in other populations.

Note: All recommendations are category 2A unless otherwise indicated.  
Clinical Trials: NCCN believes that the best management of any cancer patient is in a clinical trial. Participation in clinical trials is especially encouraged.

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HBOC-1

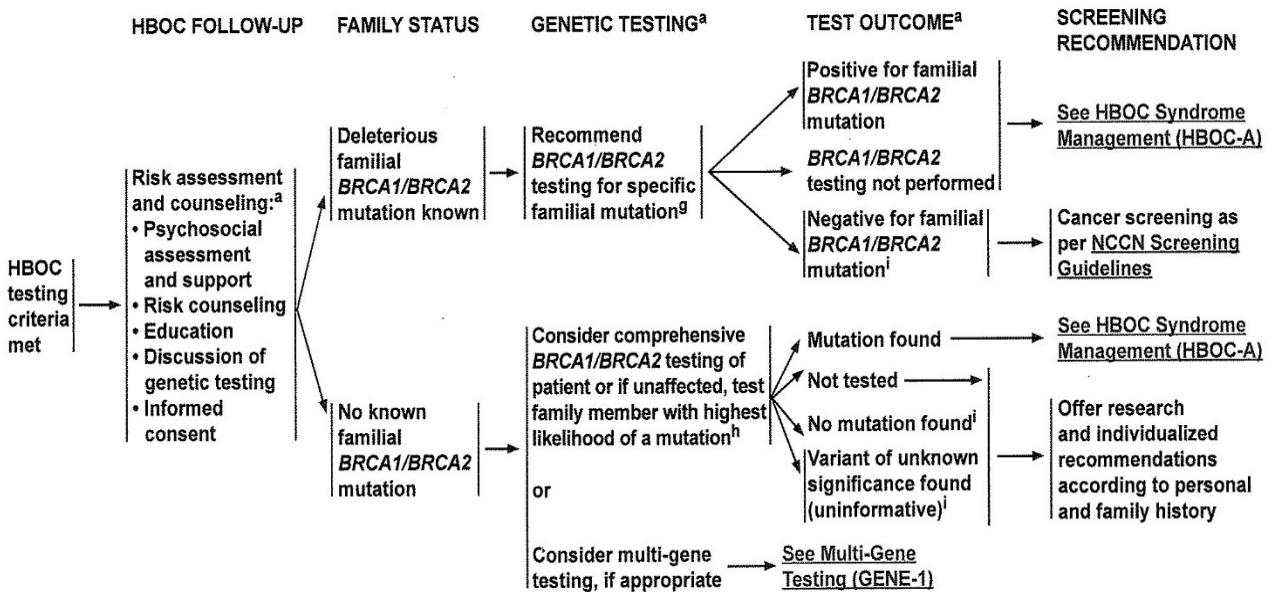
# Summary of Evidence-based Clinical Practice Guidelines:

## Genetic Susceptibility Testing for Hereditary Breast and Ovarian Cancer Syndrome

### NCCN

(continued)

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<sup>a</sup>For further details regarding the nuances of genetic counseling and testing, see [BR/OV-A](#).

<sup>g</sup>If of Ashkenazi Jewish descent, in addition to the specific familial mutation, test for all three founder mutations. Additional testing may be indicated if there is also a significant family history of cancer on the side of the family without the known mutation.

<sup>h</sup>For both affected and unaffected individuals of Ashkenazi Jewish descent with no known familial mutation, first test for the three common mutations. Then, if negative for the three mutations and ancestry also includes non-Ashkenazi Jewish relatives

or other HBOC criteria are met, consider comprehensive genetic testing. For both affected and unaffected individuals who are non-Ashkenazi Jewish and who have no known familial mutation, comprehensive genetic testing is the approach, if done.

<sup>i</sup>If no mutation found, consider testing another family member with next highest likelihood of having a mutation and/or other hereditary breast/ovarian cancer syndromes such as Li-Fraumeni ([LIFR-1](#)) and/or Cowden syndrome ([COWD-1](#)) or multi-gene testing ([GENE-1](#)). For additional information on other genetic mutations associated with breast/ovarian cancer risk for which genetic testing is clinically available, see [ADDIT-1](#).

Note: All recommendations are category 2A unless otherwise indicated.  
Clinical Trials: NCCN believes that the best management of any cancer patient is in a clinical trial. Participation in clinical trials is especially encouraged.

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HBOC-2

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## Hereditary Breast and Ovarian Cancer Syndrome

Hereditary breast and ovarian cancer (HBOC) syndrome is a condition that greatly increases a person's chance of getting certain types of cancer. HBOC syndrome is *inherited*. This means it runs in families. It is caused by a potentially harmful change in a gene that can be passed from a parent to his or her children. Most cases of HBOC syndrome result from changes in the genes called *BRCA1* and *BRCA2*.



Most breast and ovarian cancers are not related to HBOC syndrome. In fact, only about 5 of every 100 breast cancers and 10 of every 100 ovarian cancers are caused by the condition. A genetic test can tell if your cancer was caused by the altered genes associated with HBOC syndrome.

### WHY IS IT IMPORTANT TO KNOW ABOUT HBOC SYNDROME

When people have HBOC syndrome, there is a 50/50 chance that their children, sisters, and brothers could also have it. Their parents and other blood relatives (grandparents, aunts, uncles, nieces, and nephews) also are more likely than others to have the condition.

A person with HBOC syndrome has up to an 8 in 10 chance of getting breast cancer, compared to only 1 in 10 for the general population. For ovarian cancer, HBOC increases risk to as much as 4 in 10, compared to about 1 in 100 for the general population.

### SIGNS THAT HBOC SYNDROME MAY RUN IN A FAMILY

Some signs that HBOC Syndrome may run in a family are:

- Close blood relatives with breast or ovarian cancer
- Female blood relatives who got breast cancer before age 50
- A female blood relative who had breast and ovarian cancers
- A male blood relative with breast cancer
- Persons of Eastern European (Ashkenazi) Jewish ancestry have higher risk

### GENETIC COUNSELING AND TESTING FOR HBOC SYNDROME

If HBOC is suspected, talk to a genetic counselor or another health care professional who has been trained to take a complete family health history and to discuss the pros and cons of genetic testing.

Genetic testing is a kind of blood test that looks for changes in genes that are potentially harmful. If test results show there is a change in a *BRCA1* or *BRCA2* gene for a person with breast or ovarian cancer, this means that the cancer may not respond to traditional therapies and that there is a high risk that the cancer will occur again. So knowing if the individual has a *BRCA* mutation informs how to treat the cancer and which preventive options to reduce future cancer risk should be discussed.

The affected person's biological relatives can also be tested. Those who test positive can then get screened for breast cancer earlier and more often. They can also be watched carefully for signs of ovarian cancer. This could lead to finding cancer early and treating it successfully. In addition, options like preventive surgery and use of medications to prevent cancer can be considered.

Family members who did not inherit the genetic mutations associated with HBOC syndrome still can get cancer, but their chances are much lower.

[Continued on back of this page]

FOR MORE INFORMATION

First, talk with your doctor or other health care provider. More information on breast and ovarian cancer, genetic counseling services, and cancer genetic testing can be found using the resources listed below:

**Centers for Disease Control and Prevention**

- *Breast and Ovarian Cancer and Family Health History*  
[http://www.cdc.gov/genomics/resources/diseases/breast\\_ovarian\\_cancer.htm](http://www.cdc.gov/genomics/resources/diseases/breast_ovarian_cancer.htm)
- *Bring Your Brave*  
[http://www.cdc.gov/cancer/breast/young\\_women/bringyourbrave/index.htm?s\\_cid=cancer\\_BringYourBrave\\_001](http://www.cdc.gov/cancer/breast/young_women/bringyourbrave/index.htm?s_cid=cancer_BringYourBrave_001)
- *Know: BRCA*  
<https://www.knowbrca.org/>

**Connecticut Department of Public Health**

- *Cancer Genetic Counselors in Connecticut*  
[http://www.ct.gov/dph/lib/dph/genomics/cancer\\_genetic\\_counselors\\_ct2015.pdf](http://www.ct.gov/dph/lib/dph/genomics/cancer_genetic_counselors_ct2015.pdf)

**National Cancer Institute**

- *NCI Contact Center* 1-800-4CANCER  
<http://www.cancer.gov/contact/contact-center>
- *BRCA 1 and BRCA2: Cancer Risk and Genetic Testing*  
<http://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet>

**National Institutes of Health**

- *NIH Curriculum Supplement. Understanding Cancer*  
<http://www.ncbi.nlm.nih.gov/books/NBK20362/>

**National Library of Medicine**

- *Genetics Home Reference*  
[www.ghr.nlm.nih.gov/condition/breast-cancer](http://www.ghr.nlm.nih.gov/condition/breast-cancer)





## Potential Cases of Lynch Syndrome Connecticut, 2010-2011

Cancer site	Number of cancer patients <sup>a</sup>
Colon or rectum ( $\leq$ 50 yrs. of age)	455
Colon or rectum (All ages)	3,321
Endometrium (All ages)	1,269
Multiple primary sites <sup>b</sup>	330

Source: Connecticut Tumor Registry.

<sup>a</sup> Patients who were Connecticut residents at the time of diagnosis. Invasive cancers only. Includes death certificate-only registrations.

<sup>b</sup> Primary cancer sites include colon, rectum, endometrium, ovary, pancreas, small intestine, stomach, hepatobiliary tract, and renal pelvis/ureter, with the most recent diagnoses in 2010-2011.

Please note: Data in this table are not comparable to the data published in the 2012 version of this document. This table shows the number of **patients** and not the number of tumors.

**This table contains the number of patients diagnosed during 2010-2011 who may be more likely to have the variants of the genes associated with Lynch syndrome (LS), also known as hereditary nonpolyposis colorectal cancer or HNPCC.**

### WHY THESE DATA ARE IMPORTANT FOR YOU TO KNOW

LS is an inherited disorder caused by changes in the function of DNA mismatch repair genes (*MLH1*, *MSH2*, *MSH6*, and *PMS2*), which correct errors in base pairing during DNA replication. LS is the most common cause of hereditary colorectal and endometrial cancers, and may also predispose people to several other cancers (see footnote “b” to Table above).

#### **An individual with Lynch syndrome has an increased lifetime risk of certain cancers.**

- Up to 80% risk of colorectal cancer (compared to 5-6% in the general population).
- Up to 60% risk of endometrial cancer (compared to 2-3% in the general population).
- A 16% risk of a second primary colorectal cancer within 10 years of first diagnosis.

#### **Patients with either of the following might be predisposed to colorectal cancer and other cancers by the gene mutations associated with LS.**

- Early onset colorectal cancer (diagnosed at or before 50 years of age).
- Two or more of any combination of certain primary cancers of the digestive, urinary, and female reproductive systems (see footnote “b” to Table above).

#### **Patients with either of the above diagnoses should be considered for formal risk assessment and genetic counseling and/or testing.**

- Refer patients to a health care provider who is suitably trained in cancer genetics.
- Individuals diagnosed with Lynch syndrome may have a higher risk for developing future cancers and may benefit from closer monitoring or special medical management.
- Biological relatives of these individuals also may be affected.

*Developed by the Connecticut Department of Public Health Genomics Office in partnership with the Connecticut Tumor Registry and supported by Centers for Disease Control and Prevention Cooperative Agreement #DP5355-01.*

## Lynch Syndrome

### What is Lynch syndrome?

Lynch syndrome, formerly known as hereditary non-polyposis colorectal cancer (HNPCC) syndrome, is a hereditary cancer predisposition condition caused by mutations in the *MLH1*, *MSH2*, *MSH6*, or *PMS2* genes. All of these genes are “mismatch repair genes” involved in the repair of errors made during DNA replication in preparation for cell division. Mutations in any of these genes prevent the proper repair of DNA replication mistakes. As the abnormal cells continue to divide, the accumulated errors can lead to uncontrolled cell growth and possibly cancer. In addition, mutations in the epithelial cell adhesion molecule (*EPCAM*) gene that cause inactivation of the neighboring *MSH2* gene also can cause Lynch syndrome.

### Facts about Lynch syndrome

- Approximately 1 out of 35 colon cancer patients (2% to 3%) has Lynch syndrome.
- People with Lynch syndrome have a 52% to 80% chance of developing colon cancer in their lifetimes, compared to a 5% to 6% risk for people in the general population.
- Women with Lynch syndrome have a 30% to 60% chance of developing endometrial cancer in their lifetimes, compared to a 2% to 3% risk for women in the general population.
- Individuals with Lynch syndrome have a slightly increased risk of developing stomach, ovarian, hepatobiliary, small intestine, and urinary tract cancers. (See “Who is most likely...” below for other cancers related to Lynch syndrome.)
- The average age of onset of colon cancer in Lynch syndrome patients is 42 to 61 years, compared to 71 years in the general population.

### Can patients with colorectal cancer be screened for Lynch syndrome?

- Yes. Screening tests for Lynch syndrome include microsatellite instability (MSI) testing and immunohistochemistry (IHC) testing. Either or both of these tests can be performed on a tumor from a person suspected of having Lynch syndrome.
- Such screening often is recommended before a patient is referred for genetic testing.

### Who is most likely to have an *MLH1*, *MSH2* (including *EPCAM*), *MSH6*, or *PMS2* mutation?

- The likelihood is highest in families with: a history of colorectal cancer and other Lynch-syndrome-associated cancers (i.e., endometrial, stomach, small intestine, renal pelvis/ureter, ovarian, pancreatic, biliary tract, brain, and sebaceous gland adenomas and keratocanthomas); young age of onset; and synchronous or metachronous colorectal cancer.
- Not every person in such families, however, carries a harmful *MLH1*, *MSH2* (or *EPCAM*), *MSH6*, or *PMS2* mutation, and not every cancer in such families is linked to a potentially harmful mutation in one of these genes; rather, the cancers may result from sporadic somatic mutations. Furthermore, not every person who has LS will develop colorectal, endometrial, or other LS-related cancers.

### Why is genetic counseling important?

Genetic counseling helps people better understand their risk for hereditary cancer, so they can make informed decisions about genetic testing and follow-up care.

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## Lynch Syndrome

### What does genetic counseling involve?

Genetic counseling encompasses the following services:

- Reviewing an individual's personal history and detailed family medical history.
- Assessing and explaining risk for hereditary cancers and the chance of finding a mutation through genetic testing.
- Discussing the benefits, limitations, and other possible consequences of genetic testing.
- Outlining medical implications of a positive or a negative test result.
- Determining which biological relative is most appropriate to begin the genetic testing process.
- Interpreting genetic test results, and explaining what they mean for individuals and their relatives.
- Providing referrals to experts for follow-up screening and risk management.
- Providing referrals to support resources and research opportunities (including research on genetic testing, screening, treatment, etc.).
- Discussing risks and medical management options with a patient's other health care provider(s).
- Addressing common concerns about the privacy and confidentiality of personal genetic information.

### What are the benefits of genetic testing for Lynch syndrome?

- A positive test result can bring relief from uncertainty. It helps people to make informed decisions about their futures, and allows them to take steps to reduce their cancer risk through increased surveillance or other medical and lifestyle choices.
- A positive test result may help to explain why individuals or biological relatives had cancer in the past, and, should they choose to share test results, may provide their family members with useful information.
- Those who have a positive test result may be able to participate in medical research that could, in the long run, help reduce deaths from colorectal cancer.
- A negative test result may provide a sense of relief and preclude the need for special preventive checkups, tests, or surgeries.

### What are the disadvantages of genetic testing for Lynch syndrome?

Test results may affect a person's emotions, family relationships, finances, privacy, and medical choices.

- A positive result may make a person feel anxious, angry, or depressed. Medical treatments, such as surgery to try to prevent the cancer, could have serious, long-term implications. Oophorectomy and hysterectomy are recommended and have been shown to be effective, particularly after childbearing years.
- A negative result may make people feel guilty about escaping a disease that affected a loved one. They may also get a false sense of security that they have no chance of getting cancer, when their cancer risk actually is the same as that of the general population.
- Because genetic testing can reveal information about more than one family member, the emotions caused by the results can create tension within families. The results also can affect personal choices, such as marriage and childbearing.
- Privacy and confidentiality of genetic test results are additional potential concerns. HIPAA and GINA are federal acts which address the protection of genetic information.
- Genetic testing can be expensive depending on the extent of testing. Many insurance plans cover the cost of testing for persons at high risk.

**NOTE:** *Specific indicators for genetic counseling and testing vary among professional organizations. Guidelines are not a substitute for clinical judgment. Not all clinical scenarios can be anticipated, such as when there is a limited family structure or incomplete family medical history.*

# Summary of Evidence-based Clinical Practice Guidelines:

## Genetic Susceptibility Testing for Lynch Syndrome

The criteria presented were adapted from the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group recommendations,<sup>1</sup> Amsterdam II criteria,<sup>2</sup> and the revised Bethesda guidelines.<sup>3</sup>

Individuals who meet any of the following personal or family health history criteria should be suspected as having Lynch syndrome (LS), previously called hereditary nonpolyposis colorectal cancer (HNPCC), and considered for referral to genetic counseling and testing.

### 1 INDIVIDUAL IS DIAGNOSED WITH COLORECTAL CANCER,<sup>1</sup> and preliminary tumor tissue analysis indicates either of the following:

- ◆ Lack of expression on immunohistochemistry (IHC) testing.
- ◆ Microsatellite instability (MSI).

### 2 INDIVIDUAL WITHOUT A PERSONAL HISTORY OF A LYNCH SYNDROME-RELATED CANCER<sup>2</sup> (colorectal, endometrial, small intestine, renal pelvis or ureter):

- ◆ Has three or more 1<sup>st</sup>- or 2<sup>nd</sup>-degree\* blood relatives with an LS-associated cancer, and all of the following four criteria are met:<sup>2</sup>
  - ▲ At least one relative with an LS-related cancer was diagnosed before age 50; and
  - ▲ One is a 1<sup>st</sup>-degree blood relative of the other two; and
  - ▲ At least two successive generations are affected; and
  - ▲ Familial adenomatous polyposis has been excluded in the colorectal cancer cases (if any).

### 3 INDIVIDUAL WITHOUT A PERSONAL HISTORY OF A LYNCH SYNDROME-RELATED CANCER<sup>3</sup> (colorectal, endometrial, small intestine, renal pelvis/ureter, ovarian, pancreatic, hepatobiliary, brain cancers--predominantly glioblastoma as seen in Turcot syndrome, and sebaceous gland adenomas and keratoacanthomas as seen in Muir-Torre syndrome):

- ◆ Has one or more of the following:
  - ▲ A 1<sup>st</sup>-degree blood relative with a combination of colorectal cancer and another LS-related cancer, with at least one of the cancers diagnosed before age 50.
  - ▲ Two or more 1<sup>st</sup>- or 2<sup>nd</sup>- degree blood relatives diagnosed with a combination of colorectal cancer and another LS-related cancer, regardless of their ages at diagnosis.

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\* 1<sup>st</sup>-degree = parent, sibling, or child. 2<sup>nd</sup>-degree = aunt, uncle, niece, nephew, grandparent, grandchild, or half-sibling.

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<sup>1</sup> 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.

<sup>2</sup> Vasen H F, Watson P, Mecklin J P, Lynch H T. New clinical criteria for hereditary nonpolyposis colorectal cancer (HNPCC, Lynch syndrome) proposed by the International Collaborative Group on HNPCC. *Gastroenterology*. 1999; 116: 1453-1456.

<sup>3</sup> Umar A, Boland C R, Terdiman J P, Syngal S, et al. Revised Bethesda Guidelines for hereditary nonpolyposis colorectal cancer (Lynch syndrome) and microsatellite instability. *J Natl Cancer I*. 2004; 96: 261-268.



## Lynch Syndrome

Lynch syndrome (LS) is a condition that increases a person's chance of getting cancer, especially colorectal cancer (cancer of the large bowel or rectum) at a young age (before age 50). For women, it also increases the chance of getting cancer of the endometrium (lining of the uterus or womb) and ovary.

About 3 out of every 100 colorectal cancers are caused by Lynch syndrome. Women with LS have a 30% to 60% lifetime risk of developing endometrial cancer. A genetic test can tell if your cancer was caused by genes associated with Lynch syndrome.



### CAUSES OF LYNCH SYNDROME

LS is *inherited*. This means it runs in families. It is caused by changes in genes (mutations) that can be passed from a parent to his/her children. In reproduction, each parent provides one copy of each gene, giving the resulting embryo two copies of each gene. There only needs to be one copy of the gene with the LS-associated mutation for a person to have increased risk of Lynch syndrome. So if a person has LS, there is a 50% chance that his/her child has inherited a mutated copy of an LS-related gene. The person's parents and other blood relatives are also more likely to have Lynch syndrome.

### GENETIC TESTING FOR LYNCH SYNDROME

Health care experts<sup>1</sup> recommend that every person with a new diagnosis of colorectal cancer should be offered genetic testing for Lynch syndrome.

Genetic testing is a kind of blood test that can confirm or rule out that a person has the altered genes associated with Lynch syndrome. It can tell if a person's colorectal cancer was caused by Lynch syndrome. If it was, family members may also benefit from genetic counseling and testing because of their increased risk of having inherited a gene mutation associated with LS.

### BENEFITS OF GENETIC TESTING TO FAMILY MEMBERS

Blood relatives can be tested to learn if they also have Lynch syndrome. If they have Lynch syndrome, they can get screened for colorectal cancer sooner (before age 50) and more often. The most common ways of screening for colorectal cancer are colonoscopy and sigmoidoscopy. In addition to increased screening for cancer, preventive surgery may be considered.

### OTHERS WHO MAY BENEFIT FROM COUNSELING AND TESTING FOR LS

Other people who might benefit from genetic counseling are:

- People diagnosed with colorectal cancer in the past (especially before age 50).
- People with several family members who had colorectal cancer or cancer of the uterus.

*[Continued on back of this page]*

<sup>1</sup> Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Work Group

FOR MORE INFORMATION

First, talk with your doctor or other health care provider.

More information on colorectal cancer, Lynch syndrome, cancer genetic testing, and genetic counseling services can be found at the resources listed below:

**Centers for Disease Control and Prevention**

- *Genetic Testing for Lynch Syndrome*  
[http://www.cdc.gov/genomics/gtesting/EGAPP/recommend/lynch\\_consumer.htm](http://www.cdc.gov/genomics/gtesting/EGAPP/recommend/lynch_consumer.htm)

**Connecticut Department of Public Health**

- *Cancer Genetic Counselors in Connecticut*  
[http://www.ct.gov/dph/lib/dph/genomics/cancer\\_genetic\\_counselors\\_ct2015.pdf](http://www.ct.gov/dph/lib/dph/genomics/cancer_genetic_counselors_ct2015.pdf)

**National Cancer Institute**

- *Understanding Cancer*  
<http://www.ncbi.nlm.nih.gov/books/NBK20362/>
- *NCI Contact Center* 1-800-4CANCER  
<http://www.cancer.gov/contact/contact-center>

**National Center for Biotechnology Information**

- GeneReviews
  - *Lynch Syndrome*  
<http://www.ncbi.nlm.nih.gov/books/NBK1211/>

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## STANDARD 2.3

### Risk Assessment and Genetic Counseling

**Cancer risk assessment, genetic counseling, and testing services are provided to patients either on-site or by referral, by a qualified genetics professional.**

#### DEFINITION AND REQUIREMENTS

Cancer risk assessment and genetic counseling are the processes to identify and counsel people at risk for familial or hereditary cancer syndromes. The purposes of genetic counseling are to educate patients about their chance of developing cancers, help them obtain personal meaning from cancer genetic information, and empower them to make educated, informed decisions about genetic testing, cancer screening, and cancer prevention. Identifying patients at increased risk of developing cancer because of a family history of cancer or a known hereditary cancer syndrome can have dramatic effects on early detection and cancer outcome. For this reason, cancer risk assessment and genetic counseling are rapidly becoming standards of care for patients with personal and/or family history of cancer who are at high risk of having a hereditary syndrome.

The program provides cancer risk assessment and genetic counseling on-site or by referral to another facility or community-based organization.

Cancer risk assessment and genetic counseling are performed by a cancer genetics professional who has extensive experience and educational background in genetics, cancer genetics, counseling, and hereditary cancer syndromes to provide accurate risk assessment and empathetic genetic counseling to patients with cancer and their families.

Cancer risk assessment and the potential for referral may be discussed as part of the multidisciplinary cancer conference.

Genetics professionals include people with the following:

- An American Board of Genetic Counseling (ABGC) or American Board of Medical Genetics (ABMG) board-certified/board-eligible or (in some states) a licensed genetic counselor
- An American College of Medical Genetics physician board-certified in medical genetics
- A Genetics Clinical Nurse (GCN) or an Advanced Practice Nurse in Genetics (APNG), credentialed through the Genetics Nursing Credentialing Commission (GNCC). Credentialing is obtained through successful completion of a professional portfolio review process
- An advanced practice oncology nurse who is prepared at the graduate level (master or doctorate) with specialized education in cancer genetics and hereditary cancer predisposition syndromes\*; certification by the Oncology Nursing Certification Corporation is preferred
- A board-certified physician with experience in cancer genetics (defined as providing cancer risk assessment on a regular basis)

*\*Please note, specialized training in cancer genetics should be ongoing; educational seminars offered by commercial laboratories about how to perform genetic testing are not considered adequate training for cancer risk assessment and genetic counseling.*

The Cancer Committee defines the appropriate individuals who will provide risk assessment and counseling for major cancer disease sites (such as breast and colorectal). In addition, the programs not having immediate access to formal genetic counseling services should identify resources for referral.

Cancer risk assessment and genetic counseling involve pretest and posttest counseling. At a minimum, this counseling includes the following:

# AMERICAN COLLEGE OF SURGEONS – COMMISSION ON CANCER 2012 CANCER PROGRAM STANDARDS

## Pretest Counseling

- Collecting relevant information needed to assess a patient’s personal and family medical history
  - » A three- to four-generation pedigree, including detailed medical information about the patient’s first-, second-, and third-degree relatives should be obtained. Gathering information about paternal and maternal family history, ancestry/ethnicity, and consanguinity, as available, is necessary.
- Evaluating the patient’s risk
  - » One aspect of risk assessment is discussing the absolute risk that the patient will develop a specific type of cancer or cancers based on the family history. The second aspect is the risk that the patient carries a heritable or germ line mutation in a cancer susceptibility gene.
- Performing a psychosocial assessment
- Educating the patient about the suspected hereditary cancer syndrome, if appropriate
  - » The provider reviews and discusses with the patient the cancer risks associated with gene mutations, including basic concepts such as genes and inheritance patterns and more advanced concepts of penetrance and variable expressivity and the possibility of genetic heterogeneity.
- Obtaining informed consent for genetic testing (if genetic testing is recommended).

## Posttest Counseling

- Disclosure of the results and posttest counseling include a discussion of the results, significance and impact of the test results, medical management options, informing other relatives, future contact, and available resources. The test results and interpretation will be communicated to the provider.

Guidelines and recommendations for cancer risk assessment and genetic counseling for hereditary cancer syndromes are available from the Agency for Healthcare Research and Quality (AHRQ) and [...the...] NCCN.

## SPECIFICATIONS BY CATEGORY

All programs fulfill the standard as written.

### DOCUMENTATION

The program completes the SAR [Survey Application Record].

During the on-site visit, the surveyor will discuss the process for providing cancer risk assessment and genetic counseling services either on-site or by referral.

### MEASURING COMPLIANCE

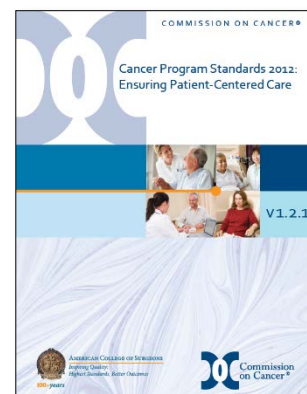
#### Rating

**(1) Compliance:** The program fulfills the following criterion:

Cancer risk assessment, genetic counseling, and testing services are provided to patients either on-site or by referral, by a qualified genetics professional.

**(5) Noncompliance:** The program does not fulfill the following criterion.

Cancer risk assessment, genetic counseling, and testing services are provided to patients either on-site or by referral, by a qualified genetics professional.



#### Reprinted from:

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>

# CANCER GENOMICS BEST PRACTICES

for Connecticut Healthcare Providers

## PROVIDER ACTION PLAN



Include the collection of family health history information as part of the patient intake process.



Review patient records and determine if appropriate genetics referral/services were provided.



Follow up with patients who may be at increased risk for hereditary cancers and have not received genetic services.



Generate referral to a genetics specialist in your area (see *Cancer Genetic Counselors in Connecticut*).



Provide appropriate management and care based on genetics consultation note and/or genetic test results.

Adapted from: Michigan Department of Community Health, Cancer Genomics Program. 2013. *Breast Cancer Genomics Best Practices for Michigan Providers*. Lansing, MI: Michigan Department of Community Health.

Developed by the Connecticut Department of Public Health Genomics Office in partnership with the Connecticut Tumor Registry and supported by Centers for Disease Control and Prevention Cooperative Agreement #DP5355-01.



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# Cancer Genetic Counselors in Connecticut

September 2015 Update

## DANBURY

### Danbury Hospital/New Milford Hospital

24 Hospital Ave., Danbury, CT 06810

Phone: (203) 739-4957 ♦ Fax: (203) 739-1922

<http://www.danburyhospital.org/en/Our-Services/Praxair-Cancer-Center/Genetic-Testing>

- Shannon Morrill-Cornelius, MS, CGC

## FARMINGTON

### UCONN Health – Carole and Ray Neag Comprehensive Cancer Center, Genetics and Genome Sciences

263 Farmington Ave., Farmington, CT 06030-2812

Phone: (860) 679-1440 ♦ Fax: (860) 679-0143

<http://humangenetics.uhc.edu/hereditary/index.html>

- Robin Schwartz, MS, CGC
- Jennifer Stroop, MS, CGC

## HARTFORD

### Hartford Hospital – Helen & Harry Gray Cancer Center

80 Seymour St., Hartford, CT 06102-5037

Phone: (860) 972-6000

<http://www.harthosp.org/cancer/BreastCancerProgram/GeneticCounseling/default.aspx>

- Sara Carroll, MS, CGC
- Kelly Genzlinger, MS, CGC
- Linda Steinmark, MS, CGC

### Saint Francis/Mount Sinai Regional Cancer Center, Karvoski Genetics Program

114 Woodland St., Hartford, CT 06105-1299

Phone: (860) 714-4679

<http://www.stfranciscare.org/Body.aspx?id=12884902378&terms=genetic%20counseling>

- Anne Wein, MS

## MERIDEN

### Midstate Medical Center – Cancer Center

435 Lewis Ave., Meriden, CT 06451

Phone: (203) 694-8631 ♦ Fax: (203) 694-7630

[https://www.midstatemedical.org/cancer\\_center.aspx](https://www.midstatemedical.org/cancer_center.aspx)

- Sara Carroll, MS, CGC

## NEW BRITAIN

### The Hospital of Central Connecticut – The Katherine Ann King Rudolph Hereditary Cancer Genetics Program

183 North Main St., New Britain, CT 06053

Phone: (860) 696-4949

<http://thocc.org/services/cancer-care/departments-services/genetic-counseling>

- Sara Carroll, MS, CGC
- Linda Steinmark, MS, CGC

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**Connecticut Department of Public Health**

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# Cancer Genetic Counselors in Connecticut

– CONTINUED –

September 2015 Update

## NEW HAVEN

### Yale New Haven Hospital - Smilow Cancer Genetics and Prevention Program

330 Orchard Street, Suite 107/109, New Haven, CT 06511

Phone: (203) 200-4362 ♦ Fax: (203) 200-1362

**Outreach clinics:** Greenwich, Griffin, and Fairfield

<https://medicine.yale.edu/cancer/patient/specialty/genetics/about.aspx>

- Karina Brierley, MS, CGC, Chief Genetic Counselor
- Michelle Ernst, MS, CGC
- Jessica DiGiovanna, MS
- Cassy Gulden, MS, CGC
- Jennifer Doherty, MS, CGC
- Niki Lovick, MS, CGC
- Arpita Neogi, MS

## NORWALK

### Norwalk Hospital – Smilow Family Cancer Center

24 Stevens Street, Norwalk CT 06856

Phone: (203) 852-2148

<http://www.norwalkhospital.org/medical-services/cancer/smilow-family-breast-health-center/services/breast-cancer-risk-assessment-program/>

- Jessica Lipschutz, MS, CGC
- Susan Ingram, MS, CGC

## STAMFORD

### Stamford Hospital – Bennett Cancer Center

30 Shelburne Rd., Stamford, CT 06904-9317

Phone: (203) 276-7693

[http://www.stamfordhospitalcancer.com/screening/screen\\_gen.aspx](http://www.stamfordhospitalcancer.com/screening/screen_gen.aspx)

- Erin Ash, MS, CGC , Coordinator

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Per An Act Concerning Licensure for Genetic Counselors,\* effective October 1, 2015:

*As used in this section, sections 2 to 6, inclusive, of this act and section 19a-14 of the general statutes, as amended by this act: (1) "Genetic counselor" means a person who has been licensed as a genetic counselor under the provisions of sections 2 to 6, inclusive, of this act; and (2) "genetic counseling" means the provision of services to individuals, couples, families and organizations by an appropriately trained individual to address the physical and psychological issues associated with the occurrence or risk of occurrence of a genetic disorder, birth defect or genetically influenced condition or disease in an individual or a family... No person may use the title "genetic counselor", "licensed genetic counselor", "gene counselor", "genetic consultant", "genetic associate", or the designation "LGC" or make use of any title, words, letters, abbreviations or insignia that may reasonably be confused with licensure as a genetic counselor unless such person is licensed pursuant to section 3 of this act or has been issued a temporary permit pursuant to section 4 of this act.*

\* Source: <http://www.cga.ct.gov/2015/TOB/S/2015SB-00857-R02-SB.htm>

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# Additional Resources

## **RESOURCES FOR HEALTH PROFESSIONALS** **HEREDITARY BREAST AND OVARIAN CANCER**

- CDC
  - ♦ Breast and Ovarian Cancer and Family Health History
    - ▲ [http://www.cdc.gov/genomics/resources/diseases/breast\\_ovarian\\_cancer.htm](http://www.cdc.gov/genomics/resources/diseases/breast_ovarian_cancer.htm)
  - ♦ Know: *BRCA* Tool [http://www.cdc.gov/cancer/breast/young\\_women/knowbrca.htm](http://www.cdc.gov/cancer/breast/young_women/knowbrca.htm)
  - ♦ Tier 1 Genomic Applications <http://www.cdc.gov/genomics/gtesting/tier.htm>
- Genetic Services – Search for cancer genetics professionals by location using these online tools:
  - ♦ <http://www.cancer.gov/search/geneticsservices/>
  - ♦ <http://www.nsgc.org/FindaGeneticCounselor>
- National Society of Genetic Counselors Practice Recommendations: Genetic Cancer Risk Assessment and Counseling
  - ♦ <http://link.springer.com/article/10.1023%2FB%3AJOGC.0000018821.48330.77>
- NCI – Genetics of Breast and Ovarian Cancer (PDQ®)
  - ♦ <http://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq>
- NCHPEG – *Hereditary Breast and Ovarian Cancer: Is Your Patient at High Risk?* CME credits
  - ♦ <http://www.nchpeg.org/hboc/>
- NCCN – Genetic Familial High-Risk Assessment: Breast and Ovarian
  - ♦ <http://www.NCCN.org>
- Tools listed in the USPSTF Recommendation Statement, 2013
  - ♦ Ontario Family History Assessment
  - ♦ Manchester Scoring System
  - ♦ Referral Screening Tool
  - ♦ Pedigree Assessment Tool
  - ♦ FHS-7

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## **LYNCH SYNDROME**

- AHRQ – *Hereditary Nonpolyposis Colorectal Cancer: Diagnostic Strategies and Their Implications (2007)*
  - ♦ <http://archive.ahrq.gov/downloads/pub/evidence/pdf/hnpcc/hnpcc.pdf>
- Collaborative Group of the Americas on Inherited Colon Cancer
  - ♦ <http://www.cgaicc.com/>
- EGAAP Working Group – Supplementary Evidence Review
  - ♦ *DNA testing strategies aimed at reducing morbidity and mortality from Lynch syndrome*
    - ▲ <http://www.egapreviews.org/docs/EGAPPWG-LynchRev.pdf>
- Journal of Genetic Counseling
  - ♦ Identification of individuals at risk for Lynch syndrome using targeted evaluations and genetic testing: National Society of Genetic Counselors and the Collaborative Group of the Americas on Inherited Colon Cancer joint practice guideline. *J Genet Couns.* 2012 Aug; 21(4):484-93.
- NCI – Genetics of Colorectal Cancer (PDQ®)
  - ♦ <http://www.cancer.gov/types/colorectal/hp/colorectal-genetics-pdq>

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# Additional Resources

## **RESOURCES FOR CONSUMERS**

### **HEREDITARY BREAST AND OVARIAN CANCER**

- Bright Pink – Resource for young women at high risk for breast/ovarian cancer
  - ◆ <https://www.brightpink.org/>
- Centers for Disease Control and Prevention (CDC)
  - ◆ Breast and Ovarian Cancer and Family Health History
    - ▲ [http://www.cdc.gov/genomics/resources/diseases/breast\\_ovarian\\_cancer.htm](http://www.cdc.gov/genomics/resources/diseases/breast_ovarian_cancer.htm)
  - ◆ Know: *BRCA* Tool
    - ▲ [http://www.cdc.gov/cancer/breast/young\\_women/knowbrca.htm](http://www.cdc.gov/cancer/breast/young_women/knowbrca.htm)
- CT DPH – *Genetic Testing for Hereditary Breast & Ovarian Cancer: What You Should Know*
  - ◆ <http://www.ct.gov/dph/cwp/view.asp?a=3134&q=396624>
- FORCE (Facing Our Risk of Cancer Empowered) – Resource for individuals at potential hereditary risk for breast or ovarian cancer.
  - ◆ <http://facingourrisk.org>
- National Cancer Institute (NCI)
  - ◆ The Genetics of Cancer
    - ▲ <http://www.cancer.gov/about-cancer/causes-prevention/genetics>
  - ◆ *BRCA1* and *BRCA2*: Cancer Risk and Genetic Testing
    - ▲ <http://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet>
- U.S. Department of Health & Human Services (DHHS) – Surgeon General's family history tool: "My Family Health Portrait". Multiple languages available.
  - ◆ <https://familyhistory.hhs.gov/fhh-web/home.action>

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## **LYNCH SYNDROME**

- CDC – Genetic Testing for Lynch Syndrome
  - ◆ [http://www.cdc.gov/genomics/gtesting/EGAPP/recommend/lynch\\_consumer.htm](http://www.cdc.gov/genomics/gtesting/EGAPP/recommend/lynch_consumer.htm)
- CT DPH – Genetic Evaluation and Testing for Hereditary Colorectal Cancer: What You Should Know
  - ◆ <http://www.ct.gov/dph/cwp/view.asp?a=3134&q=436320>
- DHHS – "My Family Health Portrait". Multiple languages available.
  - ◆ <https://familyhistory.hhs.gov/fhh-web/home.action>
- Lynch Syndrome International
  - ◆ <http://lynchcancers.com/>
- NORD (National Organization for Rare Disorders)
  - ◆ <http://rarediseases.org/rare-diseases/lynch-syndrome/>

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# Additional Resources

## GENOMICS, FAMILY HEALTH HISTORY, AND GENETIC COUNSELING

- CDC Office of Public Health Genomics
  - ◆ <http://www.cdc.gov/genomics/>
- CT DPH Genomics Office
  - ◆ *Your Family Health History Workbook: Knowing Your Past Can Influence Your Future*
    - ▲ [http://www.ct.gov/dph/lib/dph/genomics/fhh\\_wkbk.pdf](http://www.ct.gov/dph/lib/dph/genomics/fhh_wkbk.pdf)
- Health Information Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule
  - ◆ Modifications to the HIPAA Privacy, Security, Enforcement, and Breach Notification Rules Under the Health Information Technology for Economic and Clinical Health Act and the Genetic Information Nondiscrimination Act; Other Modifications to the HIPAA Rules; Final Rule
    - ▲ <http://www.gpo.gov/fdsys/pkg/FR-2013-01-25/pdf/2013-01073.pdf>
- National Human Genome Research Institute (NHGRI)
  - ◆ Genetic Information Nondiscrimination Act (GINA)
    - ▲ <https://www.genome.gov/10002328>
  - ◆ Talking Glossary of Genetic Terms, Genetic Counseling
    - ▲ <http://www.genome.gov/Glossary/index.cfm?id=79>
- National Society of Genetic Counselors
  - ◆ *How can genetic counselors help you and your family?*
    - ▲ <http://nsgc.org/p/cm/ld/fid=175>

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### **Your family health history.**

Each family and ethnic group is genetically different and you need to work with your healthcare provider to review your family health history and investigate any preexisting conditions or risk factors that may impact your life.

### **Reduce your risk to serious health issues.**

Your family history holds key information about your past and clues to your future health. Many of your physical traits (such as eye color, hair color, and height) are inherited. So, too, are risks for certain genetic conditions and health problems such as heart disease, diabetes, and some cancers. By collecting your family's health history, you can learn what health problems you may be at increased risk for in the future and how to reduce your risks.

### **Make a plan to preserve your health.**

You can't change your genes, but you can change behaviors that affect your health, such as smoking, inactivity and poor eating habits. Another change you can make is to participate in screening tests—such as mammograms and colorectal cancer screening—for early detection of disease.

**Please use the worksheet on the following pages to map out your family health history and then talk to your healthcare provider to help you design a personalized plan for maintaining your health.**

### **Congratulations...**

You are now on your way to a better understanding of your health. Contact your healthcare provider to discuss your family health history and make a personalized plan to preserve your good health.

For more detailed information, visit [www.ct.gov/dph](http://www.ct.gov/dph) and type "family health history" in the DPH search box. Or call (860) 509-8000.



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knowing your past can influence your future

**We all have a family history of something!**  
 The Surgeon General has provided a tool to help you to create a portrait of your family's health. The following worksheet will help you organize your family tree and identify common diseases that may run in families. Use this information to fill out the online version at [www.hhs.gov/familyhistory](http://www.hhs.gov/familyhistory).

Grandmother _____ _____	Grandmother _____ _____	Grandfather _____ _____
M F	M F	M F
Aunts   Uncles _____ _____	Father _____ _____	Mother _____ _____
M F	M F	M F
Brothers   Sisters _____ _____	<b>YOU</b> _____ _____	Spouse   Partner _____ _____
M F	M F	M F
	Your Children _____ _____	_____ _____
	M F	M F

**Type of information to include:**

- Major medical conditions & cause of death (where applicable)
- Age of person at disease onset
- Age of person if/when deceased from disease
- Ethnic background

Name: \_\_\_\_\_  
 Age: \_\_\_\_\_  
 Date: \_\_\_\_\_