CANCER GENOMICS BEST PRACTICES

for Connecticut Healthcare Providers

Hereditary Breast and Ovarian Cancer Syndrome

Lynch Syndrome

CONNECTICUT DEPARTMENT OF PUBLIC HEALTH
2015
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.

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

Sincerely,

Jewel Mullen, MD, MPH, MPA
Commissioner
CANCER GENOMICS BEST PRACTICES
for Connecticut Healthcare Providers

This document was developed by the Connecticut Department of Public Health Genomics Office in partnership with the Connecticut Tumor Registry and supported by Cooperative Agreement #DP5355-01, funded by the Centers for Disease Control and Prevention. Its contents are solely the responsibility of the authors and do not necessarily represent the official views of the Centers for Disease Control and Prevention or the Department of Health and Human Services.

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State of Connecticut
Department of Public Health
Jewel Mullen, MD, MPH, MPA, Commissioner
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).

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**HBOC SYNDROME**

**USPSTF Recommendation Statement, 2013**

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.

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**LYNCH SYNDROME**

**EGAPP Working Group Recommendation Statement, 2009**

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.

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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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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/).

<table>
<thead>
<tr>
<th>TERM/CONCEPT</th>
<th>DEFINITION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autosomal dominance</td>
<td>A pattern of inheritance characteristic of some genetic conditions (e.g., Hereditary Breast and Ovarian Cancer syndrome, Lynch syndrome). &quot;Autosomal&quot; means that the gene in question is located on one of the numbered, or non-sex, chromosomes. &quot;Dominant&quot; 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.</td>
</tr>
<tr>
<td>DNA</td>
<td>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.</td>
</tr>
<tr>
<td>Gene</td>
<td>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.</td>
</tr>
<tr>
<td>Genetics</td>
<td>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.</td>
</tr>
<tr>
<td>Genetic predisposition</td>
<td>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.</td>
</tr>
<tr>
<td>Mutation</td>
<td>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.</td>
</tr>
</tbody>
</table>
Potential Cases of Hereditary Breast and Ovarian Cancer Syndrome
Connecticut, 2010-2011

<table>
<thead>
<tr>
<th>Cancer site</th>
<th>Number of cancer patients¹</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female breast (≤ 50 yrs. of age)</td>
<td>1,427</td>
</tr>
<tr>
<td>Ovary (All ages)</td>
<td>576</td>
</tr>
<tr>
<td>Male breast</td>
<td>63</td>
</tr>
<tr>
<td>Multiple primary sites² (Breast-breast or breast-ovary)</td>
<td>629</td>
</tr>
<tr>
<td>Triple negative breast cancer³</td>
<td>612</td>
</tr>
</tbody>
</table>

Source: Connecticut Tumor Registry.

¹ Patients who were Connecticut residents at the time of diagnosis. Invasive cancers only. Includes death certificate-only registrations.
² More than one cancer diagnosis, with the most recent diagnosis in 2010-2011.
³ 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.

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.
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 \textit{BRCA1} and \textit{BRCA2} genes (breast cancer susceptibility genes 1 and 2). \textit{BRCA1} and \textit{BRCA2} belong to a class of genes known as tumor suppressors. In normal cells, \textit{BRCA1} and \textit{BRCA2} help ensure the stability of the cell’s genetic material (DNA) and help prevent uncontrolled cell growth. A mutated \textit{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 \textit{BRCA1} and/or \textit{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 \textit{BRCA1} and/or \textit{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 \textit{BRCA} genetic variant are more likely than non-carriers to be diagnosed with cancer before 50 years of age.
- Men with a \textit{BRCA1} and/or \textit{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 \textit{BRCA1} or \textit{BRCA2} genes is 30% to 39%.

How common are \textit{BRCA1} and \textit{BRCA2} mutations?
- An estimated 1 in 300 to 1 in 800 individuals in the general population carries a \textit{BRCA1} or \textit{BRCA2} mutation.
- Among individuals of Ashkenazi (Eastern European) Jewish ancestry, 1 in 40 individuals carries a \textit{BRCA1} or \textit{BRCA2} mutation.

Who is most likely to have a \textit{BRCA1} or \textit{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 \textit{BRCA1} or \textit{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 \textit{BRCA1} or \textit{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.

[Continued on back of this page]
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.


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<table>
<thead>
<tr>
<th>Population</th>
<th>Asymptomatic women who have not been diagnosed with BRCA-related cancer</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Recommendation</strong></td>
<td>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. (Grade: B)</td>
</tr>
<tr>
<td></td>
<td>Do not routinely recommend genetic counseling or BRCA testing to women whose family history is not associated with an increased risk for potentially harmful BRCA mutations. (Grade: D)</td>
</tr>
</tbody>
</table>

**Risk Assessment**

- Family history factors associated with increased likelihood of potentially harmful BRCA mutations include breast cancer diagnosis before age 50, bilateral breast cancer, family history of breast and ovarian cancer, presence of breast cancer in 1 or more male family member, multiple cases of breast cancer in the family, 1 or more family member with 2 primary types of BRCA-related cancer, and Ashkenazi Jewish ethnicity. 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.

**Screening Tests**

- Genetic risk assessment and BRCA 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. Tests for BRCA mutations are highly sensitive and specific for known mutations, but interpretation of results is complex and generally requires posttest counseling.

**Treatment**

- Interventions in women who are BRCA 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).

**Balance of Benefits and Harms**

- In women whose family history is associated with an increased risk for potentially harmful BRCA mutations, the net benefit of genetic testing and early intervention is moderate.
  - In women whose family history is not associated with an increased risk for potentially harmful BRCA mutations, the net benefit of genetic testing and early intervention ranges from minimal to potentially harmful.

**Other Relevant USPSTF Recommendations**

- The USPSTF has made recommendations on medications for the reduction of breast cancer risk and screening for ovarian cancer. These recommendations are available at [www.uspreventiveservicestaskforce.org](http://www.uspreventiveservicestaskforce.org).
Summary of Evidence-based Clinical Practice Guidelines:
Genetic Susceptibility Testing for Hereditary Breast and Ovarian Cancer Syndrome

NCCN

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HEREDITARY BREAST AND/OR OVARIAN CANCER SYNDROME TESTING CRITERIA\(^a,b\)
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\(^a\) + one or more of the following:
  - Diagnosed ≤45 y
  - Diagnosed ≥50 y with:
    - An additional breast cancer primary\(^c\)
    - 21 close blood relatives\(^d\) with breast cancer at any age
    - 21 close relative with pancreatic cancer
    - 21 relative with prostate cancer (Gleason score ≥7)
    - An unknown or limited family history\(^b\)
  - Diagnosed ≥80 y with:
    - Triple negative breast cancer
  - Diagnosed at any age with:
    - 21 close blood relatives\(^d\) with breast cancer diagnosed ≤50 y
    - 22 close blood relatives\(^d\) with breast cancer at any age
    - 21 close blood relative\(^d\) with invasive ovarian\(^d\) cancer
    - 22 close blood relatives\(^d\) with pancreatic cancer and/or prostate cancer (Gleason score ≥7) at any age
    - A close male blood relative\(^d\) with breast cancer
    - For an individual of ethnicity associated with higher mutation frequency (eg, Ashkenazi Jewish) no additional family history may be required\(^d\)
- Personal history of invasive ovarian\(^d\) cancer
- Personal history of male breast cancer

\(^a\)For further details regarding the nuances of genetic counseling and testing, see BRCA4-A.
\(^b\)For the purposes of these guidelines, invasive and ductal carcinoma in situ breast cancers should be included.
\(^c\)Two breast cancer primaries includes bilateral (contralateral) disease or two or more clearly separate ipsilateral primary tumors either synchronously or asynchronously.
\(^d\)Close blood relatives include first-, second-, and third-degree relatives on same side of family. (See BRCAV-B)

<table>
<thead>
<tr>
<th>Individual from a family with a known deleterious BRCA1/BRCA2 mutation</th>
<th>Personal history of breast cancer(^a) + one or more of the following:</th>
</tr>
</thead>
<tbody>
<tr>
<td>HBOC testing criteria met</td>
<td>Follow-up (HBOC-2)</td>
</tr>
<tr>
<td>If HBOC testing criteria not met, consider testing for other hereditary syndromes</td>
<td>If criteria for other hereditary syndromes not met, then cancer screening as per NCCN Screening Guidelines</td>
</tr>
</tbody>
</table>

\(^e\)Includes fallopian tube and primary peritoneal cancers. BRCA1-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 FUS 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.

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 3A 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.

HBOC-1

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<table>
<thead>
<tr>
<th>HBOC FOLLOW-UP</th>
<th>FAMILY STATUS</th>
<th>GENETIC TESTING</th>
<th>TEST OUTCOME</th>
<th>SCREENING RECOMMENDATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>家族遗传史及家族肿瘤史</td>
<td>遗传检测结果</td>
<td></td>
<td>检查结果</td>
<td>临床建议</td>
</tr>
<tr>
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<td></td>
<td>检查结果</td>
<td>临床建议</td>
</tr>
</tbody>
</table>

- **HBOC FOLLOW-UP**
  - Risk assessment and counseling:
    - Psychosocial assessment and support
    - Risk counseling
    - Education
    - Discussion of genetic testing
    - Informed consent

- **FAMILY STATUS**
  - Deletions or deleterious mutations of **BRCA1** or **BRCA2**
  - Deletions or deleterious mutations of **BRCA1** or **BRCA2**
  - Deletions or deleterious mutations of **BRCA1** or **BRCA2**

- **GENETIC TESTING**
  - **BRCA1** or **BRCA2** testing for specific familial mutations
  - **BRCA1** or **BRCA2** testing for specific familial mutations
  - **BRCA1** or **BRCA2** testing for specific familial mutations

- **TEST OUTCOME**
  - Positive for familial **BRCA1/BRCA2** mutation
  - **BRCA1/BRCA2** testing not performed
  - Negative for familial **BRCA1/BRCA2** mutation

- **SCREENING RECOMMENDATION**
  - See HBOC Syndrome Management (HBOC-A)
  - Cancer screening as per NCCN Screening Guidelines
  - See HBOC Syndrome Management (HBOC-A)
  - Offer research and individualized recommendations according to personal and family history

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*
- *Bring Your Brave*
- *Know: BRCA*
  [https://www.knowbrca.org/](https://www.knowbrca.org/)

**Connecticut Department of Public Health**
- *Cancer Genetic Counselors in Connecticut*

**National Cancer Institute**
- *NCI Contact Center 1-800-4CANCER*
  [http://www.cancer.gov/contact/contact-center](http://www.cancer.gov/contact/contact-center)
- *BRCA 1 and BRCA2: Cancer Risk and Genetic Testing*

**National Institutes of Health**
- *NIH Curriculum Supplement. Understanding Cancer*

**National Library of Medicine**
- *Genetics Home Reference*
Potential Cases of Lynch Syndrome  
Connecticut, 2010-2011

<table>
<thead>
<tr>
<th>Cancer site</th>
<th>Number of cancer patients&lt;sup&gt;a&lt;/sup&gt;</th>
</tr>
</thead>
<tbody>
<tr>
<td>Colon or rectum (≤ 50 yrs. of age)</td>
<td>455</td>
</tr>
<tr>
<td>Colon or rectum (All ages)</td>
<td>3,321</td>
</tr>
<tr>
<td>Endometrium (All ages)</td>
<td>1,269</td>
</tr>
<tr>
<td>Multiple primary sites&lt;sup&gt;b&lt;/sup&gt;</td>
<td>330</td>
</tr>
</tbody>
</table>

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 (<i>MLH1, MSH2, MSH6, and PMS2</i>), 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.
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 keratocanths); 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.

[Continued on back of this page]
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.
The criteria presented were adapted from the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group recommendations, \(^1\) Amsterdam II criteria, \(^2\) and the revised Bethesda guidelines. \(^3\)

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**, \(^1\) 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**\(^2\)
   (colorectal, endometrial, small intestine, renal pelvis or ureter):
   - Has three or more 1\(^{st}\)- or 2\(^{nd}\)-degree\(^*\) blood relatives with an LS-associated cancer, and all of the following four criteria are met:\(^2\)
     - At least one relative with an LS-related cancer was diagnosed before age 50; and
     - One is a 1\(^{st}\)-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**\(^3\)
   (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\(^{st}\)-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\(^{st}\)- or 2\(^{nd}\)-degree blood relatives diagnosed with a combination of colorectal cancer and another LS-related cancer, regardless of their ages at diagnosis.

\* 1\(^{st}\)-degree = parent, sibling, or child. 2\(^{nd}\)-degree = aunt, uncle, niece, nephew, grandparent, grandchild, or half-sibling.

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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 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]
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*

**National Cancer Institute**
- *Understanding Cancer*
- *NCI Contact Center* 1-800-4CANCER
  [http://www.cancer.gov/contact/contact-center](http://www.cancer.gov/contact/contact-center)

**National Center for Biotechnology Information**
- *GeneReviews*
  - *Lynch Syndrome*
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:
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 […] 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:
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.

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.uchc.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
• 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
• 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
• 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

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.

Connecticut Department of Public Health
410 Capitol Avenue, Hartford, CT 06106
(860) 509-8000
www.ct.gov/dph
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
• Jessica DiGiovanna, MS
• Jennifer Doherty, MS, CGC
• Michelle Ernst, MS, CGC
• Cassy Gulden, 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
• 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
• Erin Ash, MS, CGC, Coordinator

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.

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_ovaeran_cancer.htm
  - Know: BRCA Tool 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.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®)

- 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

LYNCH SYNDROME


- 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

- Journal of Genetic Counseling

- NCI – Genetics of Colorectal Cancer (PDQ®)

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.
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
  - Know: BRCA Tool

- CT DPH – Genetic Testing for Hereditary Breast & Ovarian Cancer: What You Should Know

- 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
  - BRCA1 and BRCA2: Cancer Risk and Genetic Testing

  - https://familyhistory.hhs.gov/fhh-web/home.action

LYNCH SYNDROME

- CDC – Genetic Testing for Lynch Syndrome

- CT DPH – Genetic Evaluation and Testing for Hereditary Colorectal Cancer: What You Should Know

- 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/
Additional Resources

GENOMICS, FAMILY HEALTH HISTORY, AND GENETIC COUNSELING

- CDC Office of Public Health Genomics
  - [http://www.cdc.gov/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)

- 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

- National Human Genome Research Institute (NGRI)
  - Genetic Information Nondiscrimination Act (GINA)
    - [https://www.genome.gov/10002328](https://www.genome.gov/10002328)
  - Talking Glossary of Genetic Terms, Genetic Counseling

- National Society of Genetic Counselors
  - [How can genetic counselors help you and your family?](http://nsgc.org/p/cm/ld/fid=175)


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 and type “family health history” in the DPH search box. Or call (860) 509-8000.

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.
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.

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: __________________________

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