



ST. JOHN HEALTH
BREAST CARE PROGRAM

STANDARD 4.1 GENETIC TESTING

Standard of Care:

Patients who meet specific personal or family history of breast cancer criteria should be referred for genetic counseling and testing. Referrals should be made to a High Risk Breast Clinic, Medical Geneticist, Certified Genetic Counselor, or other individual with special training in breast cancer genetics. Patients appropriate for referral for counseling and testing are:

- Any patient who has a family member with a known mutation of *BRCA1* or *BRCA2*;
- Personal history of breast cancer with one or more of the following:
 - Diagnosed < 40 years of age with or without family history;
 - Diagnosed ≤ 50 years of age or diagnosed with two breast primaries with ≥ 1 close blood relative with breast cancer ≤ 50 years old and/or ≥ 1 close blood relative with epithelial ovarian cancer at any age;
 - Diagnosed at any age with ≥ 2 close blood relatives with breast and/or epithelial ovarian cancer at any age;
 - Close male blood relative with breast cancer;
 - Personal history of epithelial ovarian cancer;
 - Ethnicity associated with a higher mutation frequency (i.e., founder populations of Ashkenazi Jewish, Icelandic, Swedish, Hungarian or other) wherein no additional family history may be required;
- Personal history of epithelial ovarian cancer;
- Personal history of male breast cancer particularly if \geq of the following is present:
 - Close male blood relative with breast cancer;
 - Close female blood relative with breast or epithelial ovarian cancer;
- Family history only:
 - Close family member (includes first, second and third degree relatives) meeting any of the above criteria.

Standard of Practice:

- Physicians should be adequately educated to identify patients who are at high risk for hereditary breast and ovarian cancer syndrome.
- Once identified, physicians should be able to answer basic questions about the syndrome and testing process.
- Physicians should make referral to an appropriate High Risk Breast Clinic,

Medical Geneticist or other trained person in cancer genetics when a patient meets above criteria or for evaluation of patient risk.

- Once a high risk patient has been identified, the High Risk Clinic or Medical Geneticist should provide appropriate risk counseling, education, psychosocial assessment and support, and obtain informed consent if any testing is performed. Physicians should be able to provide supportive care to the patient recently identified with a genetic mutation. They should be able to discuss increased surveillance, chemoprevention, and prophylactic surgery, or make referral to the appropriate resources.

Benefits of the Procedure:

- Identifying women with breast cancer who are *BRCA1* and *BRCA2* mutation carriers can help manage the contralateral breast. Lifetime risk of contralateral breast cancer in some mutation carriers can be as high 64% by age 80 with 20-30% of that risk occurring in the first 5 years after diagnosis. This may help women decide on prophylactic mastectomy, chemoprevention, prophylactic oophorectomy, or increased surveillance.
- Identifying women with or without breast cancer can help identify women at high risk for ovarian cancer. The average woman's lifetime risk of ovarian cancer is <2%. Women who are mutation carriers of *BRCA1* or *BRCA2* may have a lifetime risk of ovarian cancer as high as 44 % by age 80. This can help women decide on increased surveillance, chemoprevention, or prophylactic surgery.
- Identifying individuals who are unaffected by breast or ovarian cancer can help estimate lifetime risk of breast and ovarian cancer. Unaffected women have a lifetime risk of breast cancer as high as 87%, a lifetime risk of ovarian cancer as high as 44%. Men have a lifetime risk of breast cancer as high as 6% and increased risk of prostate cancer as high as 20%. There may also be slight increased risks of melanoma and pancreatic cancer.
- Women who are positive for a genetic mutation of *BRCA1* or *BRCA2* will likely have other family members who may be affected. This helps identify other women at high risk for breast and ovarian cancer.

References:

Characteristics of hereditary breast cancer include breast cancer prior to age 40; multiple cases of breast and/or ovarian cancer in the same individual or close blood relatives, either maternal or paternal; a family member with a known mutation in a breast cancer susceptibility gene; or a clustering of breast cancer with other cancers indicative of Li-Fraumeni syndrome or Cowden syndrome. Characteristics indicative of hereditary breast/ovarian cancer syndrome in individuals with a personal history of breast cancer include early onset of the disease at any early age, Ashkenazi Jewish ancestry, any male breast cancer, and a family history of breast and/or ovarian cancer. Individuals who have only a family history of breast and/or ovarian cancer may also be at risk. For this reason, risk assessment and counseling are considered to be integral components of genetic screening for hereditary breast cancer.

Women who meet the criteria for hereditary breast/ovarian cancer syndrome should be offered the opportunity to participate in genetic counseling delivered by a team of trained professionals. (National Comprehensive Cancer Network Guidelines: Genetic/Familial High-Risk Assessment: Breast and Ovarian 2008)

The US Preventative Services Task Force (USPSTF) found fair evidence that women with certain specific family history patterns (increased-risk family history) have an increased risk for developing breast or ovarian cancer associated with *BRCA1* or *BRCA2* mutations. The USPSTF determined that these women would benefit from genetic counseling that allows informed decision making about testing and further prophylactic treatment. This counseling should be done by suitably trained health care providers. There is insufficient evidence to determine the benefits of chemoprevention or intensive screening in improving health outcomes in these women if they test positive for deleterious *BRCA1* or *BRCA2* mutations. However, there is fair evidence that prophylactic surgery for these women significantly decreases breast and ovarian cancer incidence. Thus, the potential benefits of referral and discussion of testing and prophylactic treatment for these women may be substantial.

Appropriate genetic counseling helps women make informed decisions, can improve their knowledge and perception of absolute risk for breast and ovarian cancer, and can often reduce anxiety. Genetic counseling includes elements of counseling; risk assessment; pedigree analysis; and, in some cases, recommendations for testing for *BRCA* mutations in affected family members, the presenting patient, or both. It is best delivered by a suitably trained health care provider. (US Preventative Services Task Force, 2009)

Attested that this standard was reviewed and approved by the St. John Health Breast Advisory Board on: 9/16/09

Cheryl A. Wesen

Dated: 9/24/09

Cheryl A. Wesen, MD, FACS

Medical Director, St. John Health Breast Care Program