UTSouthwestern
Simmons Cancer Center

Genetic Testing Information for Medical Oncologists

Impact of Genetic Testing on Patient Care

Genetic testing for hereditary cancer is becoming a more widely used tool in personalized medicine. Identifying a genetic mutation associated with hereditary cancer risk can be used to determine risks for future cancers and guide cancer-risk management options, as well as determine efficacy of specific therapies on an individual basis, thereby determining treatment course. Genetic testing results can also be informative for one's family members.

Germline genetic testing typically involves analysis of a blood or saliva sample to determine whether an individual has inherited a genetic mutation that is associated with a predisposition to cancer. Clarifying future cancer risks can be informative when making recommendations for cancer surveillance (e.g., age to initiate surveillance, surveillance modalities, and frequency of ongoing surveillance), or prophylactic measures such as chemoprevention or risk-reducing surgical procedures. Somatic genetic testing is typically performed on tumor tissue to assess for genetic mutations that could guide cancer treatment. In some cases, comparative somatic genetic testing of the tumor and normal tissue can elucidate cancer etiology as well.

Using Germline Genetic Testing to Guide Systemic Therapy

PARP inhibitors such as Olaparib, Talazoparib, Rucaraib, and Niraparib have either been approved by the Food and Drug Administration or are under investigation for patients with pathogenic *BRCA* gene mutations, who have breast, ovarian, pancreatic, and prostate cancers. The use of PARP inhibitors is under investigation for patients with *ATM*, *CHEK2*, *PALB2*, *RAD51D*, and *FANCA* gene mutations and may be considered. Additionally, somatic mutations in these genes may also impact treatment options.

Pembrolizumab has been approved for patients with metastatic or unresectable solid tumors exhibiting microsatellite instability (MSI-H) or mismatch repair deficiency (dMMR) that *have* progressed following prior treatment and have no alternative treatment options. Pembroluzimab may be considered for certain patients with colorectal, small bowel, gastric, pancreatic, renal, bladder, endometrial, ovarian, prostate, and breast cancers, amongst others.

Platinum-based chemotherapy has been associated with better outcomes in patients with *BRCA*-related metastatic breast cancer and other *BRCA*-related cancers.

Chemoprevention (such as Tamoxifen, Raloxifene, and aramatose inhibitors) can be used to reduce the risk of breast cancer by up to 62% in women with *BRCA1* or *BRCA2* mutations. Additionally, chemoprevention can also be used to reduce the risk of cancer in the contralateral breast for women with *BRCA1* or *BRCA2* mutations. Chemoprevention can also help reduce the risk for breast cancer in other high-risk women, such as those with mutations in other genes related to hereditary breast cancer.

Current Landscape of Testing for Hereditary Cancer

Commercial genetic testing laboratories now offer hereditary cancer genetic testing panels, which assess multiple genes at once and test selection is often guided by patient preference and personal/family history. Update genetic testing should be considered for patients who underwent testing several years ago (typically prior to 2014).

When to Consider Genetic Testing for Hereditary Cancer

Breast Cancer
- Invasive or Intraductal diagnosed < 45
- Triple-negative diagnosed < 60
- Metastatic
- Male

Ovarian Cancer

Uterine Cancer
- Diagnosed <50 years

Pancreatic Cancer

Prostate Cancer
- Intraductal or Metastatic

Colon Cancer

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- Abnormal Immunohistochemical/Microsatellite Instability
- Diagnosed <50 years

Family history of:

- Early onset cancer(s) generally diagnosed ≤50 years
- Cancer across multiple relatives and/or multiple generations
- Relative(s) with bilateral or multiple primary cancers
- Rare cancers
- The above cancers

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Insurance and Financial Options

Most insurance companies cover the cost of genetic testing, and financial assistance may be available for patients without insurance. Self-pay options are also available.

How Can I Learn More?

For information about genetic testing and genetic counseling, visit the UT Southwestern Cancer Genetics for Health Professionals site.

For additional resources on coordinating genetics counseling and testing for patients, or providing genetics services, visit the Genetic Screening and Navigation Toolkit.

To find a genetic counselor near you, visit the National Society of Genetic Counselors Find a Genetic Counselor Tool.