



Patient Navigation in Cancer Care 2.0
Guiding patients to quality outcomes™

Precision Medicine Toolkit:

A Resource Guide for the Navigator



ACKNOWLEDGMENT

Pfizer would like to acknowledge the efforts and dedication of the members of the Precision Medicine Toolkit Committee who dedicated their knowledge, time, and efforts to enhance the care of patients with cancer through the development of *Precision Medicine Toolkit: Resource Guide for the Navigator*.

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ACADEMY OF ONCOLOGY NURSE & PATIENT NAVIGATORS: MISSION AND VISION

The mission of the Academy of Oncology Nurse & Patient Navigators (AONN+) is to advance the role of patient navigation in cancer care and survivorship care planning by providing a network for collaboration and development of best practices for the improvement of patient access to care, evidence-based cancer treatment, and quality of life during and after cancer treatment. Cancer survivorship begins at the time of cancer diagnosis.¹ One-on-one patient navigation should occur simultaneously with diagnosis and be proactive in minimizing the impact treatment can have on quality of life.² In addition, navigation should encompass community outreach to raise awareness targeted toward prevention and early diagnosis, and must encompass short-term survivorship care, including transitioning survivors efficiently and effectively under the care of their community providers.³

The vision of the Academy of Oncology Nurse and Patient Navigators (AONN+) is to achieve, through effective navigation, patient-centered superior quality cancer care coordination from pre-diagnosis through survivorship/end of life. <https://aonnonline.org/mission-and-vision>



The endorsement mark certifies that the information presented in educational seminars, publications, or other resources is considered by AONN+ to be reliable and credible.

PFIZER ONCOLOGY: OUR COMMITMENT

Pfizer Oncology is a committed partner in the cancer care community, dedicated to humanity's quest for longer, healthier, happier lives. Our goal is to improve the life of every patient with cancer and positively impact all who deal with this disease. One way we demonstrate our commitment to this goal is through our support of the patient navigation movement occurring throughout the United States.

Ask your Pfizer Oncology Account Manager about Patient Navigation in Cancer Care 2.0 to support your commitment to making a difference in the lives of patients and in shaping the future of cancer care.

Additional information regarding this program can be found at www.patientnavigation.com.

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I. INTRODUCTION & FOUNDATIONS IN PRECISION MEDICINE

Oncology nurse and patient navigators are often the first to support newly diagnosed cancer patients and their families. From diagnosis, into treatment, and even into survivorship, technologies in precision medicine can significantly impact the cancer care plan. Regardless of the type of disease, genetic testing frequently is part of the care plan, either in germline testing for inherited cancer risk or in genomics and tissue biomarker testing.

A goal of navigation is to be introduced to the patient as early as possible in the cancer continuum and promptly connect them with the appropriate services. Navigators' core competencies include educating the patients about their cancer and treatment; this also may consist of guiding referrals to genetic counseling or for biomarker testing. Understanding the broad scope of cancer genetic testing will be essential for the navigator to facilitate effective discussions with the patient. Oncology navigators already excel in care coordination and patient education, and these same skills can provide a much-needed extension into navigating cancer genetics and genomics.

Developing the oncology navigator's skills as a genetic patient educator begins by building a working knowledge of precision medicine, germline and somatic testing, and how it may specifically impact the patient population they serve. They will identify the appropriate patients who require referral to a genetic professional. The screening or identification will need to occur early in the continuum to ensure the patient is directed to the appropriate genetic resources before the start of treatment. The patient may also need to be referred at the progression of treatment. Since they have frequent and ongoing communication with patients, navigators have many opportunities to support cancer care, including in genetics and genomics.

This resource guide will serve as an essential tool for Oncology Nurse and Patient Navigators (ONNs and OPNs) involved in the care coordination of patients regarding current and emerging technologies in precision medicine.

First Principles

Our expert panel and advisory board have provided guidance based on current evidence-based literature on precision medicine to the best of our ability. Precision medicine is an ever-expanding science, and any resource guide must constantly change. Terminology for precision medicine can be overwhelming and confusing for both patients and healthcare professionals, and there are no true standardized terms at this time. With the continual evolution in precision medicine, the complex knowledge required for patients and caregivers to engage as active members of the healthcare team has rapidly and dramatically increased. Clear, concise, and consistent terminology development will allow patients the ability to make informed decisions and ultimately receive optimal care.¹

For the purposes of this introductory resource for navigators, we list here the most used terms as a foundation for learning and to optimize clarity with these concepts.

A detailed glossary of terms is also included toward the end of this document.

Genetics versus Genomics

Genetics and genomics both play roles in health and disease. Genetics refers to the study of genes and the way that certain traits or conditions are passed down from one generation to another. Genomics describes the study of all a person's genes (the genome).²

Genetics

Genetics is a term that refers to the study of genes and their roles in inheritance—in other words, the way that certain traits or conditions are passed down from one generation to another. Genetics involves scientific studies of genes and their effects. Genes (units of heredity) carry the instructions for making proteins, which direct the activities of cells and functions of the body.² Examples of genetic or inherited disorders include hereditary breast and ovarian cancer syndrome and Lynch syndrome.^{3,4}

Genomics

Genomics is a term that describes the study of all a person's genes (the genome), including interactions of those genes with each other and with the person's environment. Genomics includes the scientific study of complex diseases such as heart disease, asthma, diabetes, and cancer. These diseases are typically caused more by a combination of genetic and environmental factors than by individual genes. Genomics is offering new possibilities for therapies and treatments for some complex diseases, as well as new diagnostic methods.²

Germline Mutations

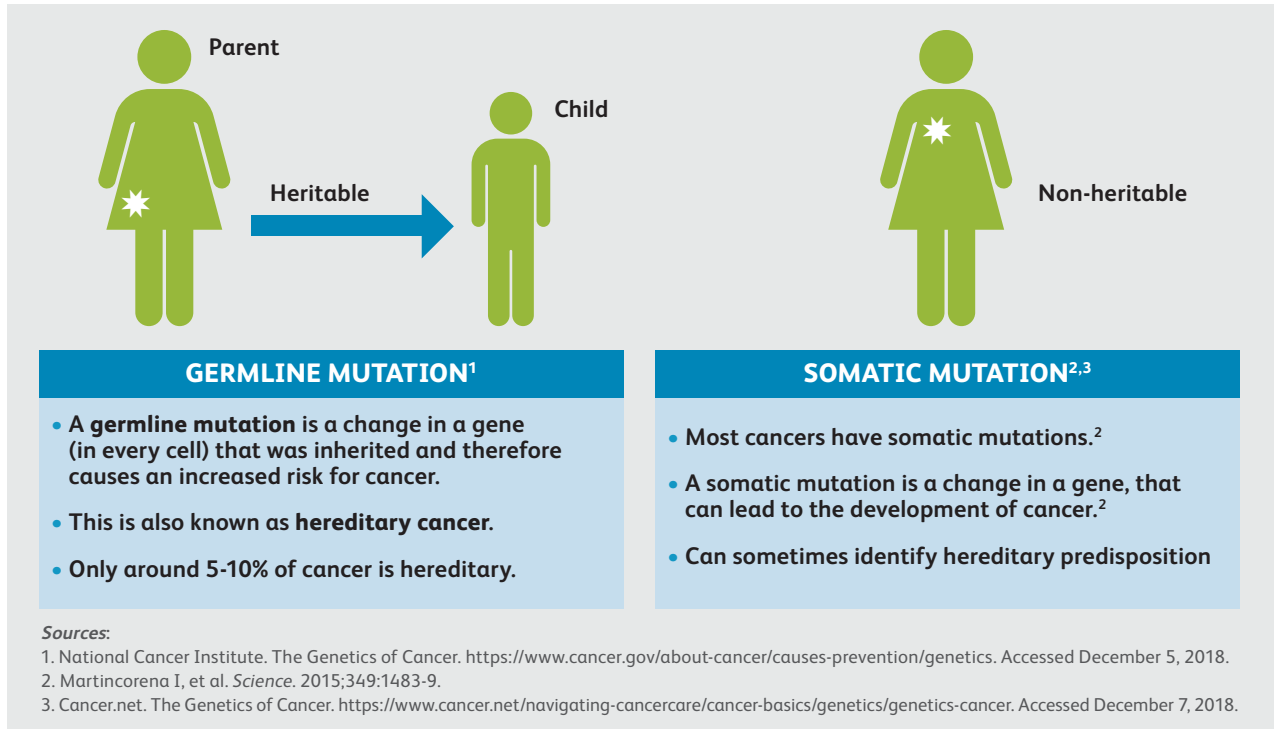
- commonly associated with Genetics, Inherited Cancer Syndrome, Cancer Genetic Testing

A gene change in a body's reproductive cell (egg or sperm) that becomes incorporated into the DNA of every cell in the body of the offspring. Germline mutations are passed on from parents to offspring and are also called germline variants (**Figure 1**).⁵⁻⁸

Somatic Mutations

- commonly associated with Genomics, Biomarker Testing, Tissue Testing

An alteration in DNA that occurs after conception. Somatic mutations can occur in any of the cells of the body except the germ cells (sperm and egg) and therefore are not passed on to children. These alterations can (but do not always) cause cancer or other diseases.⁸

FIGURE 1. Germline and Somatic Mutations⁵⁻⁸

Inherited Cancer Syndrome

- commonly associated with Genetics, Germline Mutations, Cancer Genetic Testing

A type of inherited disorder in which there is a higher-than-normal risk of certain types of cancer. Inherited cancer syndromes are caused by inherited genetic variants and may be associated with several clinical manifestations. Examples of inherited cancer syndromes include Lynch syndrome and hereditary breast and ovarian cancer syndrome.⁸

Cancer Genetic Testing

- *this is a broad term that can apply to genetics or genomics

The process of analyzing cells or tissue to look for changes in genes, chromosomes, or proteins that may be a sign of a disease or condition, such as cancer.

These changes may also be a sign that a person has an increased risk of developing a specific disease or condition. Genetic testing may be done on tumor

tissue to help diagnose cancer, plan treatment, or determine how well treatment is working.⁹ Testing can also be done via liquid biopsy to

avoid the need for tissue or when tissue is not available or sufficient for testing.



NOTES FOR NAVIGATORS

If the test is germline, the preferred general term is Genetic Testing for Inherited Risk

If the test is somatic, the preferred general term is Tissue Biomarker Testing

Biomarker Testing

- commonly associated with Genomics, Somatic Mutations, Tissue Testing

In cancer treatment, biomarker testing is a way to look for genes, proteins, and other substances (called biomarkers or tumor markers) that can provide information about cancer. Each person's cancer has a unique pattern of biomarkers. Some biomarkers affect how certain cancer treatments work.¹⁰

Biomarker testing for cancer treatment may also be called¹⁰:

- Tumor testing
- Tumor genetic testing
- Genomic testing or genomic profiling
- Molecular testing or molecular profiling
- Somatic testing
- Tumor subtyping

A biomarker test may be called a companion diagnostic test if paired with a specific treatment.¹⁰

Biomarker testing is different from genetic testing that is used to determine if someone has inherited mutations that make them more likely to get cancer. Inherited mutations are those you are born with. They are passed on to you by your parents.¹⁰

Precision Medicine

Precision medicine uses information about a person's genes or proteins to prevent, diagnose, or treat disease. In cancer, precision medicine uses specific information about a person's tumor to help make a diagnosis, plan treatment, find out how well treatment is working, or determine the prognosis. Examples of precision medicine include using targeted therapies to treat certain types of cancer, such as *HER2*-positive breast cancer cells, or tumor marker testing to help diagnose cancer, also called personalized medicine.⁹

The Navigation Process

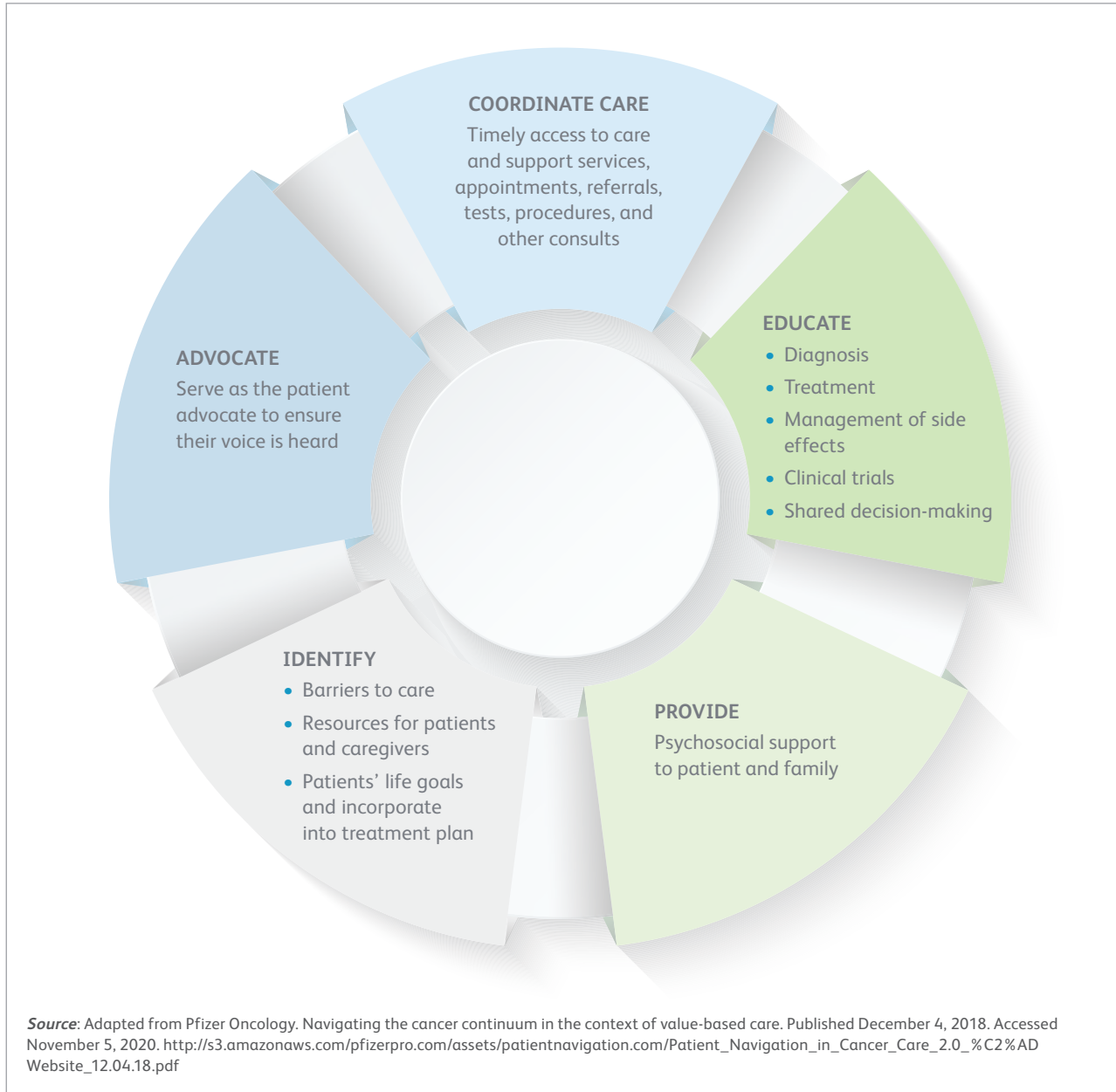
A nurse/patient navigator is defined by the Academy of Oncology Nurse & Patient Navigators (AONN+) as "a medical professional whose clinical expertise and training guides patients and their caregivers to make informed decisions, collaborating with a multidisciplinary team to allow for timely cancer screening, diagnosis, treatment, and increased supportive care across the cancer continuum."^{11,12}

A navigator's goal is to identify the earliest point of contact with the patient and their care partner/family and assess for barriers. The navigator may be one of the first members of the care team to interact with a newly diagnosed cancer patient to provide support, education, and resources, and act as the patient's advocate with the multidisciplinary team (**Figure 2**).¹³



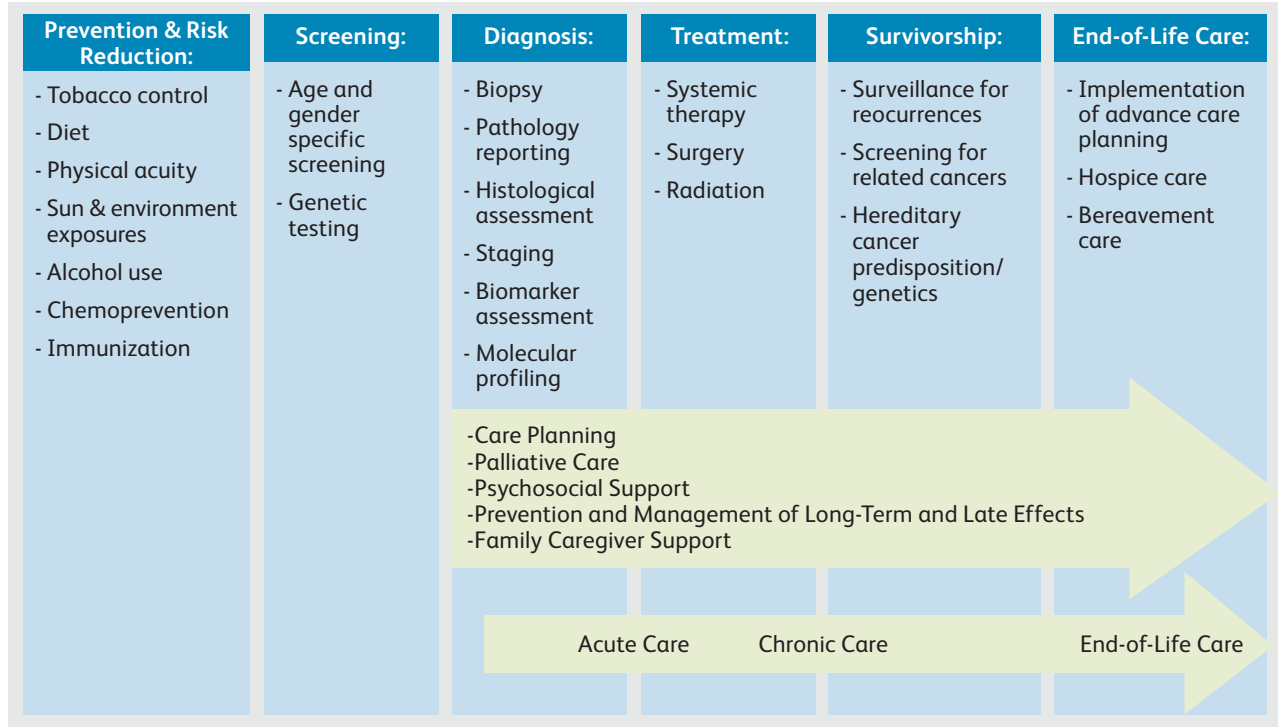
NOTES FOR NAVIGATORS

Each person's cancer has a unique pattern of biomarkers.

FIGURE 2. Goals of Oncology Navigation¹³

The National Cancer Institute describes the cancer care continuum as encompassing all phases in a patient with disease from diagnosis through treatment to the end of life.⁹

The most suitable time for the navigator to meet with the patient is early in the continuum of care (**Figure 3**). The navigator needs to stay one step ahead of the patient to facilitate timely access to care and smooth transitions across the continuum. Without the navigator assessing and providing resources and referrals to the appropriate discipline, patients may find the healthcare system overwhelming and fall through the cracks.

FIGURE 3. Domains of the Cancer Care Continuum with Examples of Activities¹³

Case Study

Navigation, the Multidisciplinary Team, and Coordination of Care

Susan is a 45-year-old woman who has been diagnosed with colon cancer. During her initial assessment, the navigator had Susan complete the National Comprehensive Cancer Network psychosocial distress screen and identify her concerns regarding her new diagnosis, causing her fear and anxiety. Susan was very worried about work and her family, especially her children. The navigator provided emotional support, education on her cancer, and requested that Susan meet with the social worker.

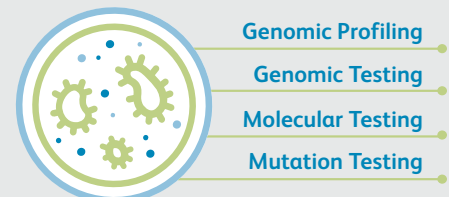
The navigator discussed her health and family history of cancer in further conversations. She realized that Susan has a significant family history of colon cancer. The navigator shared Susan's concerns and family history during the multidisciplinary cancer conference. The multidisciplinary team agreed that Susan should be referred to the genetic counselor. Results of her tumor testing and germline testing may have implications for Susan's treatment (targeted therapy and surgical recommendations), future cancer screening, and for her family (cascade testing).

The navigator was able to proactively provide the patient support, education, and referrals to the appropriate disciplines early in the continuum of care.

More on Terminology

The testing for inherited mutations is referred to as genetic testing or germline testing. The testing for somatic mutations and other nongenomic tumor characteristics can be called biomarker testing. Other terms for this are genomic testing and molecular testing, to name two.

Biomarker Testing can also be called:





NOTES FOR NAVIGATORS

So, what is the traditional role of diagnostic testing in medicine?¹⁴

- Provides information about the current or future status of the patient
- Tests are rarely used in isolation, part of a test-to-treat strategy
- Results guide clinical management
- Ideally, improve patient outcomes
- Results in a systematic approach to developing a clinical pathway

Focusing on cancer genetics and genomics, diagnostic testing takes the form mainly of biomarker testing.¹⁵ Prognostic genomic testing categorizes tumors into groups with known correlations to specific outcomes such as the likelihood of recurrence and duration of survival. This testing also sets expectations to inform treatment strategies. An example is *Oncotype DX*, which classifies the risk of recurrence for early-stage breast cancer on a 21-gene expression test and informs whether a patient would benefit from treatment.

A predictive test identifies tumors with specific molecular alterations that can be directly targeted therapeutically, guiding treatment strategy. Results of biomarker testing may also lead to the need for germline testing in patients if not done already. Examples of prognostic genomic tests are listed in the **Table**.¹⁵

TABLE. Prognostic Genomic Tests and Uses¹⁵

Tumor Type	Standard of Care Predictive Test / Drug Pairings (from NCCN Guidelines)
<i>Bladder</i>	PDL1 expression: use PD1 checkpoint inhibitors like Keytruda, Tecentriq if high
<i>Breast</i>	HER2 expression: use HER2 inhibitors like Herceptin if high ER/PR expression: use Estrogen Inhibitors, Aromatase inhibitors, CDK4/6 Inhibitors if high BRCA mutation: use PARP inhibitors like Lynparza if present Multigene expression (Oncotype test) predicts resistance to chemo
<i>Colorectal</i>	KRAS/NRAS mutation: avoid EGFR inhibitors like Vectibix or Erbitux if present Microsatellite Instability : use PD1 checkpoint inhibitors like Keytruda or Opdivo if high
<i>Gastric</i>	PDL1 expression: use PD1 checkpoint inhibitors like Keytruda, Tecentriq if high Microsatellite Instability : use PD1 checkpoint inhibitors like Keytruda, Tecentriq if high
<i>Leukemias</i>	Risk classification (based on molecular alterations) is used to select chemo regimen FLT3 mutation: use Rydabt if present IDH mutation: use IDH inhibitor like Tibsovo if present
<i>Lung (non-small cell)</i>	ALK gene fusion: use ALK inhibitors like Xalkori if present EGFR mutation: use EGFR inhibitors like Tarceva if present PDL1 expression: use PD1 checkpoint inhibitors like Keytruda, Opdivo, Tecentriq if high
<i>Melanoma</i>	BRAF mutation: use BRAF/MEK inhibitor combo like Zelboraf+Cotellic if present PDL1 expression: use PD1 checkpoint inhibitors like Keytruda, Opdivo if high
<i>Ovarian</i>	BRCA mutation: use PARP inhibitors like Lynparza if present
<i>ANY (pan-tumor approval)</i>	NTRK fusion: use TRK inhibitor Vitraki Microsatellite Instability/TMB : use PD1 inhibitor Keytruda if high

Potential Implications for Navigation Practice

Understanding how testing may impact individual patient treatment planning and follow-up for a patient with cancer can be a valuable component of the navigation team's support for patients and their families. Navigators are tasked with knowing which local genetics experts are available to service their patients. Recognizing when testing is appropriate allows navigators to connect patients to these current and emerging technologies promptly.^{13,16}

Nurse and patient navigators can play a valuable role in preparing the patient for their genetic counseling appointment in the germline testing process by participating in some or all of the following: collecting personal and family history of cancer, providing education and elements of informed consent, facilitating the process of sample collection, sharing results with the care team, and facilitating next steps for the patient and their family. In addition, navigators may also assist patients in obtaining any tumor tissue or blood biomarker testing pertinent to their diagnosis and treatment plan. They also support efforts regarding shared decision-making by discussing their specific concerns, answering questions, and helping the patient come up with questions to ask their providers about genetics and genomics.

The navigator can provide an extra layer of support, advocate for the patient and their family, and provide feedback to the genetic expert and multidisciplinary team.

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II. THE GENETICS OF CANCER

Cancer as a Genetic Inherited Disease

By nature, cancer is a genetic disease.^{1,2} The cause of cancer can be related to specific changes in genes (mutations) that regulate a cell's function, particularly in how they grow and divide. Genes contain the instructions to make proteins. Changes in these genes can lead to a misshaped protein, potentially affecting its function. If this protein is responsible for cancer-related mechanisms, such as DNA repair or tumor suppressor function, the loss of function in that protein may lead to cancer.² Genetic changes can be inherited if they are present in the germ cells; these changes are called germline mutations.²

Genetic changes can also be acquired during one's lifetime due to naturally occurring errors in cell division or exposure to carcinogens that damage DNA, such as tobacco smoke or radiation; these changes are called somatic mutations.²

The types of cancer risk can be grouped into 3 main categories: hereditary, familial, and sporadic. Hereditary cancers are associated with known germline mutations and are thought to make up 5% to 10% of all cancers.²

Sporadic cancer occurs in people with no family history of that cancer or without an inherited change in their DNA that would increase their risk for that cancer.³

Familial cancers occur in families more often than would be expected by chance. These cancers can occur at an early age and may indicate the presence of a gene mutation that increases the risk of cancer. They may also be a sign of shared environmental or lifestyle factors.³

Types of Genes Linked to Cancer

Many of the genes that contribute to cancer development fall into broad categories; this includes tumor suppressor genes and oncogenes.⁴

Tumor suppressor genes are protective genes that limit cell growth by monitoring how quickly cells divide into new cells, repairing mismatched DNA, and controlling when a cell dies.

A mutated tumor suppressor gene causes uncontrollable cell growth that may eventually lead to tumor formation. Examples of tumor suppressor genes include *BRCA1*, *BRCA2*, and *TP53*. The most commonly mutated gene in people with cancer is *TP53*. More than 50% of cancers involve a missing or damaged *TP53* gene. Most *TP53* gene mutations are acquired. Germline *TP53* mutations are rare, but patients who carry them are at a higher risk of developing several types of cancer.⁴

Oncogenes turn healthy cells into cancerous cells. Mutations in these genes are not known to be inherited. Common oncogenes include *HER2*, which is a specialized protein that controls cancer growth and spread and can be found in breast and ovarian cancer cells, among others, and the *RAS* family of genes, which makes proteins involved in cell communication pathways, cell growth, and cell death.⁴



NOTES FOR NAVIGATORS

The types of cancer risk can be grouped into 3 main categories: hereditary, familial, and sporadic.

Tumor Suppressor Genes

If a person has an error in a DNA repair gene, mistakes remain uncorrected and become mutations, which may lead to cancer, particularly mutations in tumor suppressor genes or oncogenes.⁴ Mutations in DNA repair genes may be inherited or acquired.⁴ Lynch syndrome (associated with *MLH1*, *MSH2*, *MSH6*, *PMS2*, and *EPCAM* mutations) is an example of the inherited type.⁵ *BRCA1*, *BRCA2*, and *TP53* mutations and their associated syndromes are also inherited.⁴

Because cancer is genetic in nature, there is an ever-growing number of tests that can contribute significantly to the care plan for cancer patients. Referral to the appropriate genetics professionals to get the proper tests at the right time is essential in cancer care. Navigators can best support this process based on understanding current and developing national standards for genetic counseling and testing. Navigators can also provide patients with questions to ask their providers about biomarker testing and germline testing and to see what might be appropriate for them.

National Oncology Standards for Genetic Counseling and Risk Assessment

If the program is accredited, cancer programs and navigators are versed on the Commission on Cancer (CoC) Standards and, specific to breast cancer, the National Accreditation Program for Breast Centers (NAPBC), and the National Quality Measures for Breast Cancer™ (NQMBC) 2.0.⁶⁻⁸ Even if the cancer program is not accredited by CoC and NAPBC or National Consortium of Breast Centers (NCBC), these standards represent best practice guidelines for oncology. In 2020, the CoC updated its standards,⁷ while the NAPBC updated its standards in 2018.⁸ These standards demonstrate the importance and clinical relevance of providing genetic counseling services. The navigator with this knowledge can help provide the guidance needed for a positive patient experience with the genetic counseling process.

Commission on Cancer Standard 4.4

The 2020 Commission on Cancer Standard 4.4 states that cancer programs must identify a process to provide cancer risk assessment, genetic counseling, and genetic testing services onsite or by referral.⁷ A policy and procedure must be created and include processes for the following⁷:

- Criteria for referral for a genetics evaluation
- Identification of the genetics professionals available onsite and/or by referral
- Identification of the genetics professionals qualified to perform post-test counseling either onsite and/or by referral

The individual [genetics professional] must have an educational background in cancer genetics and hereditary cancer syndromes.⁷

National Accreditation Program for Breast Centers Standard 2.16

The 2018 National Accreditation Program for Breast Centers Standard 2.16: Genetic Evaluation and Management reflects the same standards as the CoC specific for breast cancer patients. Patients with breast cancer are referred to a cancer genetics professional based on recommendations in national guidelines (eg, the National Comprehensive Cancer Network [NCCN], the American Society of Clinical Oncology [ASCO], or the American Society of Breast Surgeons). Genetic counseling is performed by a cancer genetics professional trained in genetics and cancer genetics, counseling, and hereditary cancer syndromes and who can provide accurate risk assessment and genetic counseling to cancer patients and their families.⁸

The NQNBC™ 2.0 is a quality program developed by the NCBC™.⁶ NQNBC™ standards (Genetics 1-4; Medical Oncology 7; Surgery 9) are specific standards for patients with breast cancer and provide general guidance from specific disciplines, including oncology and surgery. The NQNBC™ Program provides a way to measure performance in all or any selection of quality indicators, related to genetics.

Pre-Test Genetics Education and Access to Testing

Genetic testing can help the physician and cancer care team identify patients at hereditary cancer risk and informs the best treatment options.⁹ Germline testing results can help patients make well-informed decisions about how cancer will be treated, allowing them to be their own best advocate. It is important to have the patient ask their healthcare team about comprehensive biomarker testing when they are diagnosed and at progression.⁹ It is important to know the correct questions for the patient to ask their healthcare provider. The navigator can provide guidance and walk the patient through these questions.¹⁰

What Are You Trying to Find with Genetic Tests?

Questions to ask about genetic (germline) testing¹⁰

- Can genetic testing tell me for sure whether or not I have the condition?
- Can genetic testing be used to learn who will develop the condition in my family?
- Why should I have genetic testing?
- Are there disadvantages to having genetic testing?
- How accurate is the genetic test?
- How will I pay for the genetic test?
- Is my chance of getting an inherited condition high enough to warrant genetic testing?
- How is genetic testing done?
- How long will it take for me to receive genetic testing results?
- Who will give me the results of my genetic test, and how will I receive the results?
- Should I share genetic test results with members of my family?

The Risk for Family Members

It is ideal for the family to be made aware before the patient has germline testing; however, the patient does not have to disclose anything to their family.¹¹ Having the family participate in the upfront discussion will help with the interpretation of the results. Whether the results are positive or negative, it is crucial to consider sharing them with relatives who might also carry the mutations. If the results are positive, each first-degree relative has a 50:50 chance of testing positive for the same pathogenic mutation. Sharing information can help family members identify and manage their risk sooner. Sharing negative test results may be helpful to patients' relatives so they can be properly assessed for cancer risk by their healthcare providers, despite not needing to undergo genetic testing.¹¹



NOTES FOR NAVIGATORS

It is important to have the patient ask their healthcare team about comprehensive biomarker testing when they are diagnosed and at progression.

Ethical, Legal, and Social Implications

Many ethical, legal, and social implications associated with genetic testing and research have been raised. For genetic testing to be used safely and appropriately, these issues should be discussed with patients so that they are aware of the risks and benefits associated with testing.¹²

These potential issues can cause substantial distress for the patient and their family if not adequately addressed, ideally before testing. To protect patients from additional distress, healthcare providers should be aware of the relevant ethical, legal, and social issues related to precision medicine in healthcare. Genetic experts can help address specific patient concerns and questions regarding these issues, which include¹²:

- Communicating test results
- Direct-to-consumer tests
- Duty to disclose (warn)
- Genetic discrimination
- Informed consent
- Privacy
- Psychosocial impact
- Reproductive issues
- Societal values
- Test utility
- Test validity

Genetic testing provides comprehensive information about patients' genetic makeup as well as that of their family members, which can pose ethical and legal challenges to physicians and patients when deciding whether to disclose genetic information to family members.¹³ Failure to disclose information may lead to harm, particularly when knowledge could result in avoidance, treatment, or prevention of a genetic condition or significant changes to reproductive choices or lifestyle.^{14,15} Due to the potential harm, one may ask if there is a legal duty to warn family members about the presence of defective genes, and if so, upon whom it should be imposed.¹³



NOTES FOR NAVIGATORS

The patient should share the genetic information with¹¹:

- Close family, such as siblings or parents, preferably where the patient can answer any questions
- Children older than 18 years of age so they can make informed healthcare decisions
- Extended family, perhaps through a letter with basic information about possible risks

These family members should first connect with a genetic counselor or genetics professional to discuss testing and its possible implications.

Genetic testing can have potential emotional, social, and financial harms, including:

- Feelings of guilt: passing a pathogenic variant (mutation) on to their children or having a negative result for a pathogenic variant (mutation) present in other family members¹⁶
- The psychological stress of learning that one has a pathogenic mutation (variant) increases cancer risk and having to decide whether to share those findings with blood relatives¹⁶
- An uninformative test result, such as a report of a variant (mutation) of uncertain significance, increases uncertainty and may increase stress until results are clarified¹⁷
- Cost of testing itself and additional follow-up testing, if not covered by insurance¹⁶
- Privacy and discrimination issues¹²
- Incorrect or misleading information provided by the genetic testing¹⁷

The navigator creates a trusted relationship with a patient, and they will feel comfortable sharing their fears and concerns about genetic testing and its impact. This is an excellent opportunity for the navigator to provide support, education, and referrals to the appropriate discipline and keep the multidisciplinary team up to date.

Post-Test Genetic Education and Management

The post-test conversations about genetic testing with the healthcare team are just as important as the pre-test conversation.

Receiving genetic test results can be a highly emotional event for patients.¹⁸ The patient may experience considerable fear and anxiety prior to learning about testing results. The patient may also have a strong reaction to receiving results contrary to what they expected, even if the result is “good news.”

Post-test counseling typically involves discussion of¹⁸:

- Implications of test results, including their potential impact on management, risk reduction, screening options, prevention methods, lifestyle modifications, implications for relatives, referral to clinical trials, and support services
- Emotional impact of results and referrals for patients who need additional psychosocial support
- Dissemination plans, such as who should receive copies of results and how to discuss the results with relatives
- Provision of referrals to specialists, as needed

If applicable, the genetics professional will also determine and discuss screening and management recommendations based on the risk assessment and genetic or histology testing result. A personalized risk assessment may increase the saliency of recommendations and increase compliance.¹⁸

Some patients at increased risk may avoid or refuse enhanced or early screening or risk-reduction efforts because of denial of risk, access issues, and/or fear or uncertainty about their ability to control cancer risk. Some patients who are at reduced risk for cancer may find it difficult to accept a standard screening recommendation if they continue to perceive themselves at increased risk.¹⁸

It is vital to summarize the information provided by genetic testing and create a clear follow-up plan, including providing relevant information to family members. The genetics professional should also provide the patient with resources, such as educational materials, referrals to other health providers (if indicated), and contact information for support organizations.¹⁸

Discussing concerns and preparing the appropriate questions following genetic testing will help alleviate fear and empower patients for their follow-up visit. This is another opportune time for the navigator to educate the patient regarding the genetic counseling and testing process.



Role of Genetic Testing for Common and Rare Cancer Syndromes

As previously mentioned, sometimes the cancer is caused by an abnormal gene passed along from generation to generation.¹⁹ Often referred to as inherited cancer, it is the abnormal gene that can lead to cancer, not cancer itself. Carriers of the abnormal gene may never get a cancer diagnosis in their lifetime. So, in looking at a complete family tree, the clues to a potential inherited syndrome may be difficult to see clearly. Several factors may make it more likely that cancers in a family are caused by a family cancer syndrome, such as¹⁹:

- Many cases of the same type of cancer (especially if it is an uncommon or rare type of cancer)
- Cancers occurring at younger ages than is usual for that cancer type
- Multiple types of cancer occurring in a single person
- Cancers occurring in both of a pair of organs
- More than one childhood cancer in siblings
- Cancer occurring in the sex not usually affected
- Cancer occurring across multiple generations

Genetic Testing and Management Across the Cancer Control Continuum

Evidence-based guidelines and recommendations from a broad range of professional societies support genetic testing in certain patients. The NCCN provides guidelines that outline how to best screen for, prevent, and treat cancer, including determining who should be offered genetic testing for hereditary cancer risk and how individuals should be followed after testing.²⁰ ASCO states that recognition and management of individuals with an inherited susceptibility to cancer are core elements of oncology care.²¹



Cascade Family Testing

Cascade testing is another term that will seem foreign to the patient and their family. Cascade testing involves genetic testing in blood relatives of individuals who have been identified with specific genetic mutations.²² Testing protocols and other interventions may save lives and improve these family members' health and quality of life.

Cascade testing may include screening, counseling, or referral for a patient with a relative who has had a positive genetic test result. Screening, counseling, or referral for patients with a family history or diagnosis of cancer, but whose affected relatives have not undergone genetic testing, is not considered cascade testing.²²

The navigator as the genetic extender can provide basic education on the definition and process of cascade testing. The navigator can review who needs to be tested, who needs to be tested first, and the process of sharing information with the family.

The process begins with the identification of an individual with the condition and/or a pathogenic variant associated with the condition and then extending genetic testing to his/her at-risk biologic relatives. This process is repeated as more affected individuals or pathogenic variant carriers are identified.²³

It is vital to reach out to the family at risk for carrying the inherited cancer mutation. Cascade testing includes close relatives such as parents and children. It then extends to aunts, uncles, and cousins until all potentially at-risk relatives have been screened to ensure they do not carry the inherited cancer-risk mutation.



Prior to testing, the genetic expert will do the following to assist with the cascade testing process²⁴:

- Review genetic testing results from the patient's affected family member(s) so that the proper cascade genetic test can be ordered

- Construct a multigeneration family tree to document all relatives in the family who may be at risk for the condition
- Based on the family tree, identify all at-risk relatives, and specifically, which relatives should undergo genetic testing, or clinical screening, first
- Discuss the family's dynamics and relationships, and brainstorm ways to help with communication about the testing process, genetic testing results, or diagnoses
- Develop and provide personalized tools to share with at-risk relatives, such as family letters, family e-mails, and social media messages, and help them learn more by sharing any available condition-specific brochures
- Encourage the patient to talk with their relatives about finding genetic services for themselves

Although cancer genetic counseling and testing have been available for decades, some individuals are still hesitant and have preconceived myths. They may experience fear or concerns about what to expect for themselves or their family members. Misconceptions surrounding genetic testing and counseling could become a roadblock for valuable information that could alter their treatment plan. Here we present a couple of myths and some key information to help “bust” them.

Questions to Ask About Genomic (Somatic/Tissue) Testing

As opposed to genetic (germline) testing discussed earlier, the questions around genomic (somatic/tissue/blood) testing are different. The goal of genomic testing is to identify genomic changes in the cancer cells that are driving cancer growth for the purpose of using targeted treatments or monitoring progression of disease.²⁵ With the increasing number of available tests, knowing when and what to order can be challenging. Here are a few questions to consider with your genetics professional when weighing the option of genomic tumor testing²⁵:

- Is genomic testing appropriate for me? What would be done differently for my care?
- What is the right size panel to order? Are there limitations to larger panels?
- What kind of sample is required for genomic testing? Tissue or blood?
- When is the right time to do genomic testing in the treatment plan?

Cancer Myths and Facts

MYTH/FACT #1: Familial and genetic risk, prostate cancer example

MYTH: “As a prostate cancer patient, it doesn’t really affect my brothers, sons, or daughters with respect to cancer risk. I can’t get genetic testing because it’s probably not covered by insurance.”

FACT: Recent advances have increased the availability of genetic testing to men with advanced prostate cancer, either with or without a family history of cancer (depending on the exact clinical stage). Insurance coverage for testing is often available once formal cancer genetic counseling is completed.

Genetic testing for inherited cancer risk is often on the clinical radar for patients with breast and ovarian cancers. Genetic counseling and testing for the *BRCA1* and *BRCA2* genes have become part of the standard workup for these patients. Still, the criteria have been expanded to include prostate cancers, given the risk associated with these genes, among others. Most patients who qualify for *BRCA1* and *BRCA2* genetic testing have the option for multigene germline genetic testing, allowing for evaluating a spectrum of genes related to risks for multiple cancer types. With an identified hereditary risk through genetic testing, cascade family testing can reveal cancer risks in those without cancer that can be screened for more aggressively or even prevented with risk-reduction interventions.²⁶

With respect to advanced prostate cancer, the NCCN Guidelines support consideration of *BRCA1*- and *BRCA2*-based genetic counseling and testing for the following criteria, which can be found under hereditary breast and ovarian cancer syndrome²⁰:

- Personal history of metastatic or intraductal/criform prostate cancer, regardless of family history
- Personal history of high-grade prostate cancer (Gleason score >7 at any age)
- ≥1 close blood relatives with ovarian, pancreatic, or metastatic prostate cancer at any age or breast cancer at age <50 years; or
- ≥2 close blood relatives with breast or prostate cancer (any grade) at any age; or
- Ashkenazi-Jewish ancestry

(NCCN, 2021 – genetic/familial high-risk assessment)

Nurse navigators, treating physicians, and other healthcare team members should be aware of the new criteria, so we can recognize when cancer patients should be referred to a genetics professional (genetic counselors, advanced practice genetic nurse, or geneticist/physician specialist).

MYTH/FACT #2: Somatic tissue testing as it relates to germline testing

MYTH: “My oncologist completed the genomic testing on my tissue biopsy, and it showed a mutation in *MLH1*. I read on the internet that *MLH1* is related to Lynch syndrome, so should my family have genetic/germline testing too?”

FACT: Cancer biomarker testing can reveal pathogenic variants within genes associated with hereditary cancer syndromes but does not confirm inherited risk alone; genetic (germline) testing would be needed to confirm a syndrome, in this case, Lynch syndrome.

The differences between cancer biomarker testing for somatic mutations and germline genetic testing of blood or saliva can confuse cancer patients.^{25,27} These tests may be labeled or explained to patients as “genetic” and misconstrued as associated with inherited cancer risk. Still, they may be only a tissue-based assay geared only toward determining treatment options. Genetic (germline) testing, after genetic counseling with a specialist, would be needed to confirm an inherited cancer syndrome. Only if a pathogenic variant (mutation) is found in a genetic test (usually blood or saliva) would family cascade testing be appropriate. Hereditary cancer syndromes must be confirmed with a germline test, and cannot be found through cancer biomarker testing alone.

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III. CANCER GENETIC TESTING

Case Study #1: Localized Prostate Cancer

Mr G is a 69-year-old man who recently had a prostate biopsy due to an elevated prostate-specific antigen (PSA) score of 7.8 found at his yearly checkup.

He has met with his urologist to discuss the results, which revealed prostate cancer found in 3 of the 12 biopsy samples. His urologist explains that he has favorable intermediate-risk disease that is a small volume with a Gleason grade of 3+4. Mr G learns that his treatment options, based on the National Comprehensive Cancer Network guidelines, include surgery, radiation therapy, or active surveillance.

The patient agrees to participate in their prostate cancer multidisciplinary clinic, which includes a series of meetings with the navigator, urologic surgeon, and radiation oncologist. Mr G is considering active surveillance versus surgery at this point, but he is not quite sure which, given he is a good candidate for either choice.

Results from the biopsy, including tumor-based molecular testing, indicated low risk and tests for changes in certain genes.

He decides to proceed with active surveillance. Mr G shares that his sister died of ovarian cancer, and she did not have genetic testing for inherited risk.

Referral to the cancer genetics counselor was completed.

Family history:

- He has 2 sisters, a brother, and several nieces/nephews
- His sister with ovarian cancer has a son in his 20s
- His daughters are both in their 30s
- He is of Northern European descent, no Ashkenazi-Jewish heritage

BRCA1/2 panel-based testing was completed on a saliva sample. It showed a mutation in the *BRCA1* gene and a variant of unknown significance (VUS, a variation in a genetic sequence for which the association with disease risk is unclear) in the *PALB2* gene.

Follow-up cascade family genetic counseling/testing was recommended for his available first-degree relatives, as well as his deceased sister's son.

BRCA1 mutations have been found in one of his daughters and a nephew.

- His daughter was referred to the breast surgical oncologist to discuss high-risk screening and risk reduction options for breast and ovarian cancers.
- His nephew will start breast cancer screening at age 35, and he will consider prostate cancer screening as early as age 40. His future children may consider genetic counseling/testing as early as age 18.
- He plans to continue active surveillance for now (PSA checks every 6 months; digital rectal exam yearly; consider annual multiparametric MRI and repeat biopsy).
- There are no clinical interventions/high-risk pancreatic cancer screening based on the VUS found in the *PALB2* gene.

Cancer Biomarkers and Biomarker Testing

Cancer biomarkers include structural changes within the genome, abnormal features of gene products, or biochemical effects of the tumor. Cancer biomarkers are used for many different aspects of cancer care. Some cancer biomarkers are used to assess a person's chances of developing cancer. Other cancer biomarkers are used for early detection (screening), identification (diagnosis) of cancer, and to assess treatment response.¹

Biomarker testing is a method of looking for genes, proteins, and other substances (biomarkers or tumor markers) that can provide information about cancer. Each person's cancer has a unique pattern of biomarkers. Some biomarkers affect how certain cancer treatments work.²

Tissue-Based Biomarker Testing

Tissue-based biomarker testing is the gold standard for biomarker testing. Tissue may be secured through a biopsy or surgery and sent to the laboratory (onsite at the hospital or offsite at a testing company).¹ For some cancer types, such as lung cancer, performing a biopsy may be challenging for the patient and interventionalist alike, so a repeat biopsy to secure adequate high-quality tissue may be needed. Another way tissue may be utilized is if it was stored at the time of the initial biopsy/surgery, and there is an adequate quantity to perform biomarker testing.

Blood-Based Biomarker Testing

Blood-based biomarker testing (also known as a liquid biopsy) can identify somatic biomarkers in the blood in the form of circulating tumor DNA (ctDNA).¹ In light of recent technological advances, blood-based biomarker testing has emerged as a powerful diagnostic tool that can be performed in parallel with traditional tissue-based testing, offering valuable information on biomarkers in the patient's cancer to guide treatment decisions. Current recommendations are to utilize blood-based biomarker testing with a clinically validated next-generation sequencing platform both at the point of diagnosis and at disease progression to ensure testing for all actionable biomarkers, including somatic and non-genomic markers relevant to the patient's cancer type.^{1,3}

Making Informed Treatment Decisions Based on the Complete Biomarker Testing Results

The patient and provider must wait for their complete results from biomarker testing for all guideline-recommended biomarkers before making a treatment decision. Unless the provider feels the patient has serious symptoms, waiting 3 to 4 weeks for the results from the testing can be beneficial to the cancer patient's understanding of the type of cancer they have, which will inform the best shared decision-making with the provider on the treatment options. Navigators can assist patients and family members in understanding why waiting, while nerve-racking and frustrating, is essential for treatment selection.



NOTES FOR NAVIGATORS

Cancer biomarkers include structural changes within the genome, abnormal features of gene products, or biochemical effects of the tumor.

After Getting Biomarker Testing, the Patient Should Ask⁴:

- What are the results of these tests?
- How will the results affect my treatment?
- The test results are negative: should I be retested?
- The test results are not clear: should I be retested?
- Are there any medications that target my type of cancer?
- Are there any clinical trials open to me based on these results?
- Will I need these tests again? If so, why? When?
- How can I get a copy of the report with my biomarker testing results?

Future Directions and Emerging Technologies in Cancer Genomics

Most genetic assays will likely be based on liquid biopsy in the future.⁵ This technique can identify tumor markers in the blood in the form of circulating tumor cells (CTCs), ctDNA, or other circulating biomarkers. CTCs can be shed from tumors into the bloodstream, while ctDNA can be released by tumor cells due to cell death or active processes.

Resources available for navigators to use in gathering new information for patients:

Cancer Risk Assessment Checklist⁶

- Simple family history screening tool
- Patient talking points
- Assessing genetic services tool
- Professional society guidelines
- Provider educational resources
- Patient education materials

Application/Principles of Genetic Testing as the First Step in Patient Access to Precision Medicine

Precision medicine will allow us, in many cases, to standardize care by identifying which patients are most likely to respond to a specific treatment based on a precise diagnosis of their condition.⁷

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IV. APPROACHES TO IMPROVE THE PROCESS OF IDENTIFICATION, TESTING, MANAGEMENT, AND REFERRAL OF INDIVIDUALS WHERE GENETIC TESTING IS APPROPRIATE

Make Discussion About Genetic Testing a Part of Your Routine

- Talk about genetic testing with your patients and their providers
 - Print a pocket-sized list of the cancers that warrant a serious discussion regardless of family history
- Collect 3-generation family history
 - Submit this on behalf of your patients to your precision medicine resources—it will save patients from needing to do it!
- Set up a mechanism for a referral for genetic testing
 - Triage complex cases
 - The collaborative relationship for case-sharing
- Talk about cascade testing
 - Know the process for your patient's relatives
 - www.nsgc.org—find a genetic counselor

The navigator has many opportunities to assess and educate the newly diagnosed cancer patient for precision medicine resources. These opportunities may be at the initial case presentation at the tumor conferences, new patient multidisciplinary center visit, new referral patient intake process, or progression of disease, just to name a few.

Brief family history and identification of hereditary cancers would include¹:

- **Family history of cancer:** Having 3 or more relatives on the same side of the family with the same or related forms of cancer.
- **Cancer at an early age:** Having 2 or more relatives diagnosed with cancer at an early age. This factor may differ depending on the type of cancer.
- **Multiple cancers:** When one relative develops 2 or more types of cancer.
- **Rare cancers:** Some types of cancer, such as ovarian cancer, adrenocortical cancer, or sarcoma, are linked to inherited genetic mutations.

Genetic tests are available for some types of cancer. These include the following¹:

- Breast cancer
- Ovarian cancer
- Colon cancer
- Thyroid cancer
- Prostate cancer
- Pancreatic cancer
- Melanoma
- Sarcoma
- Kidney cancer
- Stomach cancer

A small pocket-sized genetic testing resource list would be an excellent addition to the assessment tools utilized by the navigator and the healthcare team. The guide should include cancers that warrant a serious discussion regardless of family history. These individuals need to be referred for genetic services.

The family health history is a record of diseases and conditions that run in the family. The family members may share genes, habits, and environments that can affect your patient's cancer risk.²

What Information Is Needed?²

Gather information about the patient and their family members (grandparents, parents, aunts and uncles, siblings, and children).² The information collected should include all instances of cancer (who had it and what kind, the age of the patient when they were diagnosed, whether the relative is still alive, and if not, what was the cause of death).

A fundamental principle of navigation is to empower the patient in their care; this can be done by providing a simple family history form for your patient and their family to complete with instructions. The navigator can review the document before the genetic testing visit. If the navigator has time allotted, they can assist the patient with completing the tool. This will help the genetic expert tremendously, and the outcome will be a productive visit.

Referrals to the Cancer Genetic Expert

Included in the definition of navigation is to remove barriers and provide access to medical and psychosocial care; the navigator has an obligation to get the patient to the correct services in a timely manner. Simple genetic referral guidelines in oncology and an easy referral process must be established. The navigator will also need to refer complex cases as soon as possible based on the established guidelines (ie, National Comprehensive Cancer Network Genetic/Familial High-Risk Assessment)³ and follow-up with the patient to discuss the next steps. Communication will be vital in keeping the entire healthcare team up to date on the patient's care plan.

Barriers to Genetic Testing in Cancer Care

There are quite a few barriers to oncology genetic testing for the patient, healthcare system, and healthcare team.⁵⁻⁸ These barriers can include lack of knowledge, genetic literacy, fear of the results, access to care, timeliness of referrals, workforce shortages, and insurance coverage, just to name a few.

Patient Issues

There are barriers and concerns with access to genetic testing for oncology patients. From the patient perspective, they may be^{9,10}:

- Experiencing fear, anxiety, and psychological distress due to the process
- Finding the terminology overwhelming
- Realizing that the results or uncertainty of the test results is having a negative impact on the patient and their family

The navigator can provide support and basic education on genetic testing. Lack of appropriate insurance coverage is also a concern for the patient. The cost of care is a top concern for the cancer patient, ranging from no coverage to high copays and deductibles and out-of-pocket expenses.⁵ It is illegal to discriminate based on insurance; there are protections in place. The patient's goals with genetic testing must be discussed; for example, what are the patient's treatment goals? What are the patient's life goals?



NOTES FOR NAVIGATORS

The navigator has an obligation to get the patient to the correct services in a timely manner.



NOTES FOR NAVIGATORS

The multidisciplinary team led by the physician should refer the following patients to a cancer genetic expert⁴:

- Cancer was diagnosed at an unusually young age
- Several different types of cancer occurred in the same person
- Cancer in both organs in a set of paired organs, such as both kidneys or both breasts
- Several first-degree relatives (the parents, siblings, or children of an individual) have the same type of cancer (eg, a mother, daughter, and sisters with breast cancer); family members with breast or ovarian cancer; family members with colon cancer and endometrial cancer
- Unusual cases of a specific cancer type (eg, breast cancer in a man)
- The presence of birth defects that are known to be associated with inherited cancer syndromes, such as certain noncancerous (benign) skin growths and skeletal abnormalities associated with neurofibromatosis type 1
- Being a member of a racial or ethnic group that is known to have an increased risk of having a certain inherited cancer susceptibility syndrome and having 1 or more of the above features as well
- Several family members with cancer

Healthcare Team Issues

From the genetic expert and multidisciplinary healthcare team perspectives, there is a lack of access to genetic experts in oncology.¹¹ There may be no services onsite at the cancer center or untrained staff providing the genetic testing and counseling services.⁶ Many new organizations also provide offsite genetic testing and a more recent concept of direct-to-consumer testing, which may or may not include counseling, a vital component of genetic testing. The referral process or lack of consistent referrals is a concern for the counselors, as well as poor quality or incomplete documentation of family history.

Provider concerns exist, such as:

“These syndromes are so rare, and I almost never find a positive result.”

“It won’t affect how I manage this patient’s care.”

“My patients don’t want to know if they have a bad gene.”

“I don’t have time to get into this with my patients.”

“I am worried about how complicated testing is and which test(s) to order.”

“Insurance will not cover this expensive test, and the patient will be left with high out-of-pocket costs.”

Healthcare System Concerns

The healthcare system shares some of the same concerns as the patients and counselors, such as lack of insurance and qualified personnel. There may also be an inadequate referral process or a lack of referrals to genetic experts. The providers may lack knowledge of the importance of genetic testing. There are also still some negative attitudes and misperceptions about genetic testing.⁵

Communication and Decision Aids

Navigators know the importance of clear and concise education and communication regarding cancer and treatment. Patient empowerment is also a goal for navigation programs. Encouraging the patient to feel confidence in their discussions and ensure their questions and concerns are heard with their healthcare team is necessary. The communication on genetic testing needs to be at a level that the patients understand and incorporate their learning style. Always start from what the patient already knows; for example, ask the patient basic questions such as, what do you know about genetic testing? What has your doctor told you about the genetic testing process? After this initial discussion, the navigator can provide additional detail and ask the patient to repeat their understanding. This process needs to be repeated until the patient fully understands what genetic testing is and the process. Patient educational materials and decision aids need to be utilized to reinforce the conversation. These materials need to include a glossary of terms at the fifth-grade level or below,¹² describing the process from referral to the genetic expert to receiving the results.

According to 2019 Health Center Program data, 43% of health centers were capable of providing telemedicine, compared with 95% of the health centers that reported using telehealth in 2020 during the coronavirus disease 2019 (COVID-19) pandemic.¹³ Telehealth can facilitate access to care, reduce the risk for transmission of SARS-CoV-2 (the virus that causes COVID-19), conserve scarce medical supplies, and reduce strain on healthcare capacity and facilities while supporting continuity of care.

Some of the benefits of telehealth include¹⁴:

- Comfort and convenience
- Control of infectious illness
- Better assessment
- Family connections
- Primary care and chronic conditions management

Other options for genetic consultation include initial visits via the patient portal, telephone, virtual visits, and video chats.

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V. PUTTING IT ALL TOGETHER: THE ROLE OF GENETIC EXTENDER FOR NURSE & PATIENT NAVIGATORS

Proposed role and value of navigators as a genetic extender

As mentioned earlier, the genetic extender is not a new role or an additional position at your cancer center; these tasks can be incorporated into the skill set of the navigator. The navigator's goal is to meet the patient and their family at the earliest point of entry in the continuum, and their role is to coordinate care across the continuum. Since the navigator is introduced early in the patient's cancer journey, they are in the perfect position to start the initial education about genetic testing, discuss patient concerns, collect family history, and review the genetic counseling and testing process. This additional support for the genetic expert facilitates timely access to services that will enhance the patient experience and treatment decisions.

Role delineation within the multidisciplinary cancer care team

As with any new process, it is essential to have the multidisciplinary team meet to review roles, responsibilities, and processes. The team would include the genetic expert(s), leadership, physician, key stakeholder, oncology nurse or patient navigator, and clerical support if available. The role delineation exercise will ensure that each staff member understands their responsibilities and the patient flow for the genetic counseling process and testing. A patient or nonclinical navigator would have responsibilities different from the clinical navigator. The genetic extender may include members of the cancer care team such as clinical and nonclinical patient navigators, medical assistants, and clinic registered nurses, just to name a few. The professional staff needs to function to the top of their license. They may be overwhelmed by tasks that could be completed by a nonclinical staff member, thus the importance of discussing role delineation with the multidisciplinary team. Appropriate training includes formal genetic education as well as on-the-job training.

The definition and core competencies for both clinical and nonclinical navigators are available through the Oncology Nursing Society (ONS) and the Academy of Oncology Nurse & Patient Navigators (AONN+).¹ It is vital that the navigators, regardless of their role, are allotted time to perform these additional genetic extender responsibilities. The oncology nurse and patient navigator must participate in the appropriate genetic counseling education to function as a genetic extender.

Additional responsibilities for the nonclinical navigator in the genetic extender role would include:

- Early identification and triage of at-risk patients based on guidelines provided by the genetic expert and key stakeholders
- Scheduling appointments, tests, or procedures as ordered by the provider
- Requesting the appropriate medical records for the genetic counseling visit
- Providing basic patient education that has been reviewed/approved by the genetic counselor and key stakeholders, including basic terminology, the role of the genetic expert, and the biomarker testing and counseling process

Additional responsibilities for the clinical navigator in the genetic extender role would include:

- Early identification and triage of at-risk individuals based on national guidelines
- Review of the patient's medical records to initiate the family history assessment
- Contacting the patient to get any information that was not included in the medical record for the family history
- Genetic patient assessment and education
- Getting informed consent from the patient

Demonstrating Value Added to the Practice/Healthcare System**Metrics, Downstream Revenue**

Metrics demonstrate the success and sustainability of an oncology program and are important to enhancing the care provided to cancer patients. Some examples of genetic program metrics include:

- Time from cancer diagnosis to genetic expert appointment
- Time from referral to a genetic expert to the genetic counseling appointment
- Support services provided after the genetic referral
- Downstream revenue as a result of the genetic expert referral

The act of genetic counseling has positive downstream revenue effects, including the professional genetic counselor and the genetic extender. The return on investment (ROI) is significant, and ROI or business performance should be included as a metric for the genetic program.

There are significant opportunities for downstream revenue for cancer genetic experts. In a recent study over a 10-year period, the downstream revenue generated from cancer genetic expert identification of patients with hereditary breast and ovarian cancer (HBOC) and Lynch syndrome (LS) was \$32.79 million in US dollars (USD) (mean/year = \$3.25 million USD and mean/patient = \$77,000 USD).² One full-time genetic expert would generate \$1.49 million to \$1.86 million USD in revenue per year (\$1.26 million-\$1.58 million USD for HBOC-positive patients and \$227,000-\$284,000 USD for LS-positive patients per year).

Glossary of Educational Terms That Can Be Shared with Patients³:

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Precision Medicine

Precision medicine is a newer way to find the proper treatment for each patient, based on the cancer subtype. Previously, the only option was to treat all cancers of one type (such as lung or breast cancer) with the same treatment. Instead, doctors use biomarker testing to find your cancer subtype in precision medicine. These tests show which treatment is likely to work best for you. Precision medicine is only available for certain types and stages of cancer.

Biomarker

A biomarker is a sign of disease or abnormal function that can be measured in your blood, tissue, or bodily fluid. In cancer, biomarkers are often used to help choose the best treatment for you. These biomarkers can be proteins, genes, or gene mutations.

Biomarkers are often referred to by a 3- or 4-letter and/or number abbreviation. Examples of biomarkers are *HER2* in breast cancer or *EGFR* in lung cancer.

Biomarker Testing

Biomarker testing helps your doctor match the right drugs to the specific subtype of cancer you have. In biomarker testing, a sample of your cancer is collected from your blood, bodily fluids, or tissue taken during surgery or biopsy. Your sample is sent to a lab. The test looks for biomarkers in your cancer sample. The test results can be used to help guide your treatment options. Biomarkers tell your doctor about the subtype of cancer in your body.

Mutation in the Cancer (Acquired Mutation)

Two kinds of mutations can be found in cancer cells—mutations you inherit and mutations you acquire during your lifetime. Inherited mutations are found in all your cells. Acquired mutations may only be found in the cancer's cells. Acquired mutations occur as you get older and result from the wear and tear of life. These mutations were not inherited and cannot be passed on to children. Mutations in the cancer can affect how the cancer grows and spreads. These mutations can also define the cancer's subtype. Mutations in the cancer can be a type of biomarker. A biopsy sample or liquid biopsy is needed to test for mutations in the cancer.

Acquired mutations are one type of genetic mutation. These mutations develop over your lifetime. These mutations occur as you get older and are the result of the wear and tear of life. You do not inherit acquired mutations from your parents, and you cannot pass these mutations on to children. These mutations are not found in all your cells. There may be acquired mutations in just the cancer cells. The cancer's genes may be tested for acquired mutations. Acquired mutations can affect how the cancer grows and spreads.

Testing the Cancer for Mutations

Comprehensive Biomarker Testing, Cancer Marker Testing

One specific type of biomarker testing looks for mutations in the cancer. Biomarker testing helps your doctor match the right drugs to the specific subtype of cancer you have. This may be a targeted therapy drug or immunotherapy. In biomarker testing, a sample of your cancer is collected from your blood, bodily fluids, or tissue taken during surgery or biopsy. Your sample is sent to a lab. The test looks for biomarkers in your cancer sample. The test results can be used to help guide your treatment options. Biomarkers tell your doctor about the subtype of the cancer in your body.

Inherited Mutation

A genetic mutation is a change in a gene. Mutations in your genes are inherited from your parents and can be passed on to children. Your genes may be tested for mutations that increase your risk of getting cancer.

Genetic Testing for Inherited Cancer Risk

Testing for people who have not been diagnosed with cancer. In some cases, cancer runs in families. This means an increased risk of cancer is passed down from parents to their biological children. You may have heard this called "inherited cancer." What is inherited is a gene mutation (a change in your genes) that increases your cancer risk. It does not pass down the cancer itself. Testing to see if you inherited a mutation that increases your risk of getting cancer when you have no diagnosis of cancer can be called "genetic testing for inherited cancer risk."

Genetic Testing for an Inherited Mutation

Testing for people who have been diagnosed with cancer. In some cases, cancer runs in families. It seems to be passed down from parents to children. However, what is inherited is a mutation (change) in your genes that increases your risk of cancer. It is not the cancer itself that is inherited.

Genes

Genes carry the information that passes on traits inherited from parents to children. Chemically, a gene is made of DNA. Genes affect the way our cells and bodies work.

Cancer Types

The type of cancer you have is usually named for the organ or tissues where the cancer forms. Common cancer types are lung, breast, colorectal, prostate, and skin cancers.

Targeted Therapy Drugs

Targeted therapy drugs keep cancer from growing and spreading with less harm to cells that are not cancer. These drugs “target” specific cancer subtypes. They are only likely to work in those specific subtypes.

These drugs can work in several ways:

- They can find cancer cells.
- They can destroy cancer cells directly.
- They can stop cancer cells from growing uncontrollably.
- Or they can cut off the blood supply that tumors need to grow and survive.

Solid Tumor

Solid tumor cancers are cancer types that begin in organs or tissue, not in blood cells. These kinds of cancers often develop tumors, which are masses of abnormal tissue. Examples of solid tumor cancers are:

- Lung cancer
- Breast cancer
- Colorectal cancer
- Skin cancer
- Prostate cancer

Blood Cancer

Blood cancer begins in blood cells, in the bone marrow, or in the cells of the immune system. Blood cancers do not usually form tumors. Examples of blood cancers are:

- Leukemia
- Lymphoma
- Myeloma

Liquid “Biopsy”

This is a biomarker test done through bloodwork. It tests tumor DNA or tumor cells found circulating in your blood. Your doctor may choose to do a liquid “biopsy” since it only needs a sample of blood. If a liquid “biopsy” comes back with a positive result, your doctor can use those results to choose treatment.

Tissue Biopsy

When cells or tissues are removed and sent to a lab to see whether cancer is present. Biopsies require surgery or a needle to remove the cells or tissues. Samples from tissue biopsies can be used to test for biomarkers in the cancer.

