



Empowering Personalized Care with Genetic and Genomic Testing

Over the last several decades, groundbreaking developments in genetics technology have come to the forefront of cancer prevention, diagnosis and treatment. Genetic testing can now identify inherited genetic mutations that increase an individual's risk of cancer, aiding in the development of preventative and treatment strategies, while genomic testing can identify mutations in tumors that may respond to specific therapeutic agents, enabling more targeted treatments. By having a basic knowledge of genetic and genomic testing, physicians can better understand which patients may benefit from these advanced testing technologies.

Genetic Testing: Is Your Patient Predisposed to Cancer?

Genetic testing can determine whether or not a patient has an inherited disease-related gene condition or genetic abnormality that may increase the risk of getting cancer. More than 50 hereditary cancer syndromes have been identified¹ and genetic testing is available for many of them, including those linked to breast, ovarian, pancreatic, melanoma, and colon cancers.

Researchers estimate as many as 10 percent of all cancers may be associated with an inherited mutation that contributes to cancer development.²

Who Should Have Genetic Testing?

When considering whether to recommend genetic testing to a patient, physicians should determine the patient's personal and family history to see if certain cancers or patterns of cancer are present. Genetic counseling and testing should be considered for patients who meet any of the following criteria:³

- Family member with multiple primary cancers.
- Several relatives on one side of the family who have had the same type of cancer.
- Several cancers in the family linked to a single gene mutation (such as breast, ovarian, colon or pancreatic cancers).
- Family or personal history of early onset cancer (usually before age 50).
- Relatives with a rare cancer, such as breast cancer in a male or retinoblastoma.
- Ashkenazi Jewish ancestry with breast or ovarian cancer at any age.
- A physical finding that is linked to an inherited cancer, such as colon polyps.
- Family members with a known genetic mutation.

How Does Genetic Testing Impact Patient Care?

Genetic testing can help physicians provide better care by enabling the implementation of risk mitigation strategies beyond the standard of care for high-risk patients. More aggressive screening may be indicated for certain genetic cancer syndromes, increasing the odds of detecting cancer early when treatment is most effective. Physicians can also use the information to help patients make informed decisions about medications, preventative surgery, diet, exercise and other lifestyle changes that may reduce the risk of developing cancer.

Hereditary gene mutations not only affect the patient, but may also impact the patient's entire family, as these genes can be passed on to children by both the father and the mother. Consequently the patient's parents, siblings and children could also carry the mutation and be at a higher risk for cancer. If the test results are shared with family members, they may want to also be tested to enable them to make better decisions about their own healthcare.

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Genomic Testing: Bringing Precision Medicine to Cancer Care

Genetics and genomics are frequently used interchangeably, but they are not the same. While genetics focuses on single genes and the role they play in passing conditions or traits from one generation to the next, genomics examines mutations in tumor genes that may determine cancer behavior. Genomic information about cancer provides better diagnoses and treatment strategies tailored specifically to patients' tumors, an approach called precision or personalized medicine.

Guiding More Targeted Treatments

With genomic testing, the genomic makeup of abnormalities, or mutations, within the cancerous tissue can be identified. These abnormalities may drive cancer growth, determining how aggressive the cancer is or whether or not it spreads to other parts of the body. By identifying the genetic profile of a patient's tumor, a more precise treatment may be available. If the mutation corresponds to one already identified, a targeted therapy may exist that attacks only that specific mutation without harming normal cells, making it more effective and less likely to be toxic to patients compared to traditional treatment options.

"We can use new innovations in genetic testing to more precisely predict, diagnose, and treat conditions in individual patients, instead of having to settle for a one-size-fits-all approach."

*- American Medical Association President
Dr. Andrew W. Gurman.⁴*

Recent Advancements in Personalized Cancer Care

In recent years, drugs have been developed to fight cancer by specifically responding to genomic changes in the cancer cells. This can include blocking enzymes that contribute to the abnormal growth of cancer cells, blocking abnormal gene expression of cancer cells, or inhibiting molecular signaling pathways that go into overdrive in cancer cells.⁵

A National Research Council report on precision medicine demonstrates the role genomic testing can play in advancing cancer care by contrasting breast cancer treatment today and in the past. Twenty-five years ago, few treatment options existed for the disease, and treatment usually involved hormone therapy or chemotherapy, both of which may have side effects that impact quality of life. Today, there are many targeted treatment options for breast cancer based on specific markers in the patient's tumor, providing more effective treatments with fewer side effects.⁶ Cancer genomics research has also defined the molecular taxonomy for various cancer types and subtypes based on their genetics, enabling a more precise diagnosis and personalized treatment strategy for patients.⁵

The Genetic Risk Evaluation and Testing Program

Southern Cancer Center is pleased to offer cancer risk assessment services as part of the comprehensive quality care we provide. We encourage physicians in our community to contact us to learn more about genetic and genomic testing, or for assistance in determining which patients may benefit from these advanced testing technologies.

Sources

1. <https://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet#q3> 2. <https://www.asco.org/about-asco/press-center/news-releases/asco-releases-updated-policy-statement-genetic-and-genomic> 3. <https://www.cancer.org/cancer/cancer-causes/genetics/understanding-genetic-testing-for-cancer.html> 4. <http://theweek.com/articles/639296/how-genomics-dramatically-changing-future-medicine> 5. <https://www.cancer.gov/about-nci/organization/ccg/cancer-genomics-overview> 6. <https://www.cancer.org/latest-news/personalized-medicine-redefining-cancer-and-its-treatment.html>

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