

BACKGROUND

Many terms are used to describe cancer biomarkers in patient education materials and literature for healthcare professionals. One of the greatest areas of confusion centers on the use of multigene panels designed to identify germline or somatic changes in DNA. The inconsistent use of terminology may confuse patients when clinicians discuss the importance of biomarker testing and review test results.



HEREDITARY (GERMLINE) TESTING: genetic testing that may reveal a hereditary cancer syndrome or may identify patients who may be eligible for certain types of targeted therapy.



BIOMARKER OR TUMOR (SOMATIC) TESTING: often performed using a multigene panel and/or other tests (eg, protein expression) to identify patients who may be eligible for targeted therapy and/or immunotherapy. Testing is usually performed on tumor tissue or blood.

TIP



Be Clear About Somatic vs Germline Testing

The Consistent Testing Terminology Working Group convened in 2020 and 2021 to recommend ways to reduce confusion when healthcare professionals speak with patients about biomarker tests and their results.¹ They recommend harmonizing language and simplifying communication by using the terms:

- **Biomarker testing** to discuss tests that identify characteristics, targetable findings, or other test results originating from malignant tissue or blood, and
- **Genetic testing for an inherited mutation** and **genetic testing for inherited cancer risk** for tests that identify germline mutations (also known as variants in the genetics community).

TIP



Explain the Importance of Germline Testing in Patients with Cancer

In 2022, the American Society of Clinical Oncology (ASCO) released a publication titled “Somatic Genomic Testing in Patients With Metastatic or Advanced Cancer: ASCO Provisional Clinical Opinion.”² The document stated that germline testing “should NOT be limited by family history–based or clinical criteria used for familial risk assessment.”² Rather, germline testing for genetic alterations linked to approved therapies “should be performed in patients with metastatic or advanced solid tumors considered for such treatment.”² Therefore, it is important to emphasize to the patient that germline testing will not only help inform cancer risks for patients and their relatives—it could also help guide optimal treatment recommendations.

TIP



Remind Patients That Somatic Tests May Reveal Certain Germline Variants

Remind patients that somatic biomarker tests are not designed to identify germline variants; however, certain variants (eg, BRCA) could be germline in nature.³ Some laboratories perform paired somatic-germline testing (also called matched tumor-normal testing) to identify variants that are somatic or germline.⁴ Confirmatory testing at a laboratory with clinically-validated diagnostic testing for hereditary cancer syndromes should be performed when a germline variant is suspected on a somatic test report.⁵

 **TIP**

Be Clear About What Is Being Tested

Include answers to the following types of questions along with explanations of different cancer biomarker tests and their results:

- What is being tested? Tumor tissue, normal tissue, blood, or something else?
- Is the test looking for an abnormality in the tumor or in the rest of the body?
- Why is the test being performed, and how will results impact care (including treatment options or inclusion in clinical trials)?
- Will these results have meaning or impact for family members?

 **TIP**

Be Clear About What Test Results Mean

When testing results are available, be clear when reviewing:

- How the results will be used to guide immediate treatment decisions
- Whether the results provide information about prognosis and/or future cancer risk
- How the results of germline testing (if performed) may impact family members
- Whether these tests may need to be repeated as treatment continues (eg, to evaluate disease progression or identify new treatment options).

 **TIP**

Connect Patients with Genetic Counselors

Remind patients that the results of ongoing research are revealing greater understanding about variants in cancer genes. Certified genetic counselors (CGCs) can provide valuable support and explain the significance of test results.⁶ They may also help to facilitate cascade testing of at-risk family members and to provide personalized medical recommendations for managing elevated cancer risks based on test results and family history. CGCs can also assess whether patients may face financial barriers to testing and refer patients to financial advocates or patient assistance resources.

 **TIP**

Remind Patients That Misinformation Remains Pervasive

Much of the existing lay and professional literature may interchange terms like variant and mutation or attempt to oversimplify complex biological concepts. As a result, misinformation or confusing information may be found, especially on the internet. Remind patients that public information may be oversimplified, outdated, inaccurate, or that it may include different definitions for certain terms.

 **TIP**

Consider New Roles Like Precision Medicine Stewards

Some cancer programs are creating new roles such as precision medicine stewards or biomarker navigators to ensure timely and appropriate testing. These individuals may have backgrounds in areas such as nursing, genetics, or laboratory science and can play an important role in identifying and addressing barriers so that all eligible patients are appropriately tested. Precision medicine stewards and biomarker navigators may also communicate test results to patients and explain how the information may inform treatment decisions.

Learn more: acc-cancer.org/cancer-diagnostics

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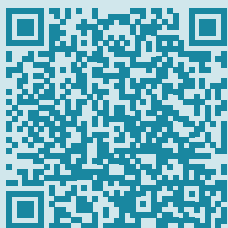
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