Biomarker Tests and Cancer Treatment

Cancer biomarkers are genes, proteins, or other substances that can be tested for to reveal important details about a person’s cancer. There are many known cancer biomarkers, and new ones are being found all the time.

Testing for biomarkers might not be helpful for everyone. But for people with many types of cancer, biomarker testing might provide useful information that could affect their treatment options.

If a person has been diagnosed with cancer, biomarker testing may show whether:

- The cancer is more likely to grow and spread
- Certain cancer treatments are likely (or unlikely) to be helpful
- The cancer treatment is working

Biomarker testing is useful in other ways too, including:

- Helping to determine if a person might be at higher risk for some types of cancer
- Showing if someone might have a certain type of cancer (although a biopsy is typically needed to know for sure)
- Looking for possible signs of cancer returning

Depending on why and how it’s done, biomarker testing for cancer treatment can have different names. If your health care team recommends any of the following, they are recommending tests for biomarkers:

- Tumor testing, tumor genetic testing, tumor marker testing, or tumor subtyping
- Genomic testing, genomic profiling, or genome sequencing
• Molecular testing or molecular profiling
• Somatic testing
• Next generation sequencing

A biomarker test might also be called a **companion diagnostic test** if it’s used to help determine if a specific medicine might be helpful.

### Why biomarker tests can be helpful for treatment

Not all cancer cells are alike. Even among people with the same type of cancer (such as breast or lung cancer), the cancer cells can have different **gene changes** or different levels of certain proteins that can help the cancer cells grow. These changes might also affect how well the cancer responds to certain types of treatment, such as **targeted drug therapy** and **immunotherapy**, which work best when the cancer cells have certain features that make them different from normal cells. Some of these differences can be tested for as biomarkers.

The differences in the features of cancer cells are the basis for how targeted drugs work. Targeted drugs are often designed to attack cancer cells that have certain gene or protein changes. Biomarker testing - that is, checking a person’s tumor cells for these changes - before treatment can help show if these drugs are likely (or unlikely) to be helpful.

For example, in some types of cancer, people whose cancer cells have certain changes in the *EGFR* gene can often be helped with targeted drugs known as **EGFR inhibitors**. Biomarker testing can be done before treatment to find out if a person’s cancer cells have an *EGFR* gene change. This can show if an EGFR inhibitor might be helpful in treating their cancer.

Cancer medicines aren’t equally effective for all people, regardless of whether they were designed to work against certain biomarkers. As doctors have looked for ways to tell which people might be helped by different treatments, more biomarkers have been discovered that help tell when certain treatments might work.

For example, drugs called **immune checkpoint inhibitors** can often help the body’s immune system attack cancer cells. These drugs can be very helpful in treating some cancers, but they don’t work against others. Since these drugs were first approved, research has shown that they’re more likely to work if a person’s cancer cells have a lot of gene changes (mutations) that make them different from normal cells. Some biomarker tests can look for signs of these changes, such as the cells having a high
tumor mutational burden (TMB), or the cells having changes that turn off one of the DNA mismatch repair (MMR) genes (which normally help limit DNA mutations).

Some biomarker tests can help show if a person might benefit from more treatment, but they don’t specify if a particular treatment would be helpful.

For example, some tests look at patterns of genes (or proteins) in cancer cells to see if the cancer is more likely to return after the initial treatment. If it is, then further treatment might be useful. But if not, a person could be spared from more treatment and its possible side effects.

Biomarker testing isn’t helpful yet for all types of cancer. But as more is learned about what causes cancer cells to grow and as new cancer treatments are developed, biomarker testing will likely become even more important in the future.

**Biomarker testing and precision medicine**

Biomarker testing is an important part of precision medicine, also called personalized medicine. Precision medicine is an approach in which medical care is tailored based on the specific genes, proteins, and other substances in a person’s body.

Cancer treatment is typically based on where the cancer starts in the body (such as the breast, colon, lung, prostate, etc.), as well as other factors such as if the cancer has spread. But precision medicine goes beyond choosing treatment based on just the type and extent of the cancer. Precision medicine uses biomarker and other tests to choose treatments that are most likely to be helpful for each person, while at the same time sparing them from treatments that aren’t likely to help.

The idea of precision medicine isn’t new. In fact, some treatments based on biomarker testing have been around for decades. But in recent years, as researchers have come to better understand many of the different changes in genes, proteins, and other substances that can make cancer cells grow and spread, they’ve developed many newer cancer treatments and biomarker tests.

While progress is being made every day, precision medicine still isn’t part of routine care for people with many types of cancer. This will likely change in the future, as researchers learn more about what makes each type of cancer grow and how this growth can be stopped.

**Types of biomarker tests for cancer treatment**
Different types of biomarker tests can be done to help determine the best cancer treatment options:

- Many cancer biomarker tests look for gene changes in the cancer cells, while some tests measure certain proteins or other kinds of markers.
- Some tests look at a single biomarker. Other tests check for many biomarkers at the same time (such as patterns of certain genes or proteins). Some tests can even look at all of the genes inside cancer cells.
- Many biomarker tests are for people with a certain type of cancer. But some tests look for biomarkers that can be found in many different cancer types.
- Many biomarker tests are done on tumor samples removed during a biopsy or surgery. But some biomarker tests can be done on samples of blood or other body fluids. (See “How is biomarker testing done?”)

**Do I need biomarker testing for my cancer?**

If you have cancer, talk with someone from your health care team about whether biomarker testing should be part of your care.

For people with certain types of cancer, biomarker testing is done routinely to help guide treatment decisions. For other types of cancer, it might only be done for people who need systemic treatments (medicines), such as if the cancer has spread or is no longer responding to other treatments. And for some types of cancer, biomarker testing might not yet be part of routine care.

Here are some examples of biomarker tests that might help guide treatment decisions for some of the more common types of cancer:

- **Non-small cell lung cancer:** Changes in genes such as *KRAS, EGFR, ALK, ROS1, RET, MET,* and *BRAF*
- **Breast cancer:** Estrogen receptor (ER) and progesterone receptor (PR) proteins; HER2 gene or protein status; changes in genes such as *BRCA1, BRCA2,* and *PIK3CA*
- **Colorectal cancer:** Changes in genes such as *KRAS, NRAS,* and *BRAF*
- **Melanoma skin cancer:** Changes in the *BRAF* gene
- **Any cancer:** Changes in *NTRK* genes; changes in mismatch repair (MMR) genes; levels of microsatellite instability (MSI); tumor mutational burden (TMB)
This is by no means a complete list. Some people might not need all of the biomarker tests for their type of cancer, and some people might benefit from getting other tests not listed here. People with other types of cancer might also be helped by biomarker testing. And new biomarker tests are being developed all the time. To learn more about which biomarker tests might be helpful for you, talk to your health care team, or read about your cancer type.

Biomarker testing can sometimes be complex, and not all biomarker tests are available at every cancer treatment center. Check with your health care team to see if biomarker testing is available where you get your cancer care, or if your testing might need to be done elsewhere.

If your health care team thinks biomarker testing might be helpful for you, it’s also a good idea to check with your health insurance provider to see if they will cover it. (See “How much does biomarker testing for cancer treatment cost?”)

How is biomarker testing done?

If you and your health care team decide biomarker tests are right for you, they will need to get samples of your cancer cells for testing.

- If you have a solid tumor (as opposed to a cancer of the blood), they will most likely use the samples taken during a biopsy of the tumor or during surgery. If it’s hard to get a sample for testing, sometimes a blood sample (which often contains parts of tumor cells) can be used instead. This is called a liquid biopsy.
- If you have a blood cancer (such as a leukemia), a blood or bone marrow sample might be tested.
- For some cancers, other body fluids, such as urine, might be tested.

Your samples will be sent to a lab, where they will be tested for certain biomarkers. Some cancer centers can do these tests in their own labs, but sometimes tests might need to be sent to a central lab. Once the testing is done, the lab will send back a report that lists the biomarkers in your cancer cells, as well as what this might mean about treatments that could help you. It might take a week or longer to get the results, depending on which tests are being done and if the samples need to be sent out for testing. Your health care team will discuss the results with you to help you understand your treatment options.

For some biomarker tests that look at genes, you might also need to give a sample of your healthy cells (usually from a blood or saliva sample). Tests are then done to look
for differences between the genes in your cancer cells and your healthy cells. Testing both kinds of cells can show if a change in the cancer cells is a somatic mutation (also known as an acquired mutation) that happened during your lifetime, or if it was an inherited mutation passed down from a parent. (Somatic mutations cause most cancers, and they can’t be passed on to family members.)

What do the results of a biomarker test mean?

You might get different types of results from biomarker testing. The results might show

- That your cancer has a certain biomarker that is targeted by a known treatment, so this might be an option to help treat your cancer. This may be available as an FDA-approved treatment, an off-label treatment, or by taking part in a clinical trial.
- That your cancer doesn’t have certain biomarkers, which means some treatments are unlikely to work. This could help spare you from getting treatments that won’t help.
- If you’re likely to benefit from treatment, without pointing to a specific treatment. For example, a test might show that your cancer is more likely to come back after the first treatment, so getting more treatment now could help lower this risk.
- Some changes in your cancer cells that aren’t helpful in making treatment decisions, such as a gene change that is thought to be harmless (benign) or whose effects aren’t clear (known as a variant of unknown significance, or VUS). These results might still be on the lab test report, even though they aren’t used to make treatment decisions.

Some results might lead to genetic testing

Some biomarker tests might find gene changes that you may have been born with that increase your risk of cancer (or other diseases). These changes are often called inherited or germline mutations.

If such a change is found during biomarker testing, your health care team may recommend genetic counseling to help you decide if you should consider genetic testing for an inherited mutation. The results of genetic testing could affect both you and your family members, so your team will likely recommend that you speak with someone trained in genetic counseling and testing first. They can help you understand what the test results might mean for you and your family.
When might biomarker testing not be helpful for cancer treatment?

Biomarker tests might help you and your treatment team decide which treatment(s) might be best for you. But it’s important to know that these tests don’t help everyone. Here are some reasons why biomarker testing might not help you:

- You aren’t able to get a biopsy (or surgery) to collect tumor samples for biomarker testing for some reason.
- There isn’t enough tumor tissue in your biopsy sample for biomarker testing.
- Your insurance doesn’t cover the costs of testing for certain biomarkers.
- Testing is done, but it doesn’t find biomarkers in your cancer that match up with available treatments.
- Biomarker testing identifies a treatment that is being tested in a clinical trial, but you can’t enroll in the study for some reason.
- A liquid biopsy is the only way to test your cancer for biomarkers, but it might not show any markers (even if they’re on the cancer cells) because not every cancer sheds tumor cells into the blood.

Even if your test finds a biomarker that matches an available treatment, the treatment might not work for you. Targeted drugs and immunotherapy might be more likely to work in people with certain biomarkers than other treatments, but they still don’t work for everyone.

And, as with other types of cancer treatments, there is the chance that even if a treatment is helpful, it might become less effective over time, as the cancer cells could change and become resistant to some treatments.

How much does biomarker testing for cancer treatment cost?

The cost of biomarker testing can vary widely, depending on the type of test you get, the type of cancer you have, and if you have insurance, as well as what type of insurance you have.

For people with advanced cancer, some biomarker tests are covered by Medicare and Medicaid. Private insurance companies often cover the cost of a biomarker test if there is enough proof that the test is needed to guide treatment decisions. Tests without enough proof to support their value may be considered experimental, so they might not be covered by insurance.

Biomarker testing might be done as part of clinical trials of new cancer treatments. If
you join one of these clinical trials, the cost of testing for any new biomarkers will likely be covered by the study. The study coordinator can give you more information about related costs.

**Other uses of cancer biomarkers**

While biomarkers are most often linked to cancer treatment, they can have other uses as well.

**Determining a person’s cancer risk**

Some biomarkers can be useful in helping to determine if a person is at higher risk for cancer.

For example, in people with some cancers (such as colorectal cancer and endometrial cancer), the cancer cells are typically tested for genetic changes such as microsatellite instability (MSI) and defective mismatch repair genes (dMMR). Finding these changes could affect cancer treatment options, but it could also signal that a person should consider genetic testing\(^{15}\) for Lynch syndrome, an inherited condition in which people are at higher risk for several types of cancer. People with this syndrome might benefit from screening for these other cancers. Family members might want to consider genetic testing to see if they are affected as well.

**Finding cancer**

Some cancer biomarkers can be used to try to find cancer early in people without symptoms, or to help diagnose cancer in someone who already has signs or symptoms (although most often a biopsy\(^ {16}\) is needed to know for sure).

For example, while it’s not a perfect test, the prostate-specific antigen (PSA) blood test\(^ {17}\) can be used to screen for prostate cancer, especially in older men without any symptoms. (PSA is a protein that can often be found at higher levels in the blood of men with prostate cancer.)

In men who do have symptoms that might be from prostate cancer, the PSA test can also be used to help determine if a prostate biopsy might be needed to look for cancer.

Newer lab tests\(^ {18}\) that look at levels of several different biomarker proteins can also sometimes be used to help tell if a biopsy should be done (or if it should be done again, if the first biopsy didn’t find prostate cancer but the doctor still suspects cancer because the PSA level is high).
As another example, certain gene mutations are commonly found in the cells of some other types of cancers. Finding these mutations in cells can help confirm the diagnosis of that cancer.

**Showing if treatment has been effective**

Biomarker testing can also be used during or after treatment for some cancers to see how well the treatment is working.

For example, in people with chronic myeloid leukemia (CML), the leukemia cells contain a mutated gene called BCR-ABL. Very sensitive lab tests can tell how many copies of this gene mutation are in a blood sample (which indicates how many CML cells are present). The number of copies can be measured before treatment is started, and then again sometime later to see how well the treatment is working.

**Looking for signs of cancer returning**

Some biomarker tests can be used after treatment, to look for possible signs of the cancer coming back. Both the PSA blood test in prostate cancer and testing for the BCR-ABL gene in CML are examples of tests that can be used to look for signs of cancer recurrence, often before the person is having any symptoms. This might allow treatment of the recurrence to start earlier.

**Hyperlinks**


References


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