



Biomarker testing for cancer treatment adaptable resource

Table of Contents

1. [Is biomarker testing part of precision medicine?](#)

Biomarker testing is an important part of precision medicine. For cancer treatment, precision medicine means using biomarker testing and other tests to select treatments that are most likely to help you, while sparing you from receiving ineffective treatments.

2. [What is biomarker testing for cancer treatment?](#)

Biomarker testing looks for genes, proteins, or other biological molecules in your blood, bodily fluids, or tissues to help diagnose or identify the type of cancer you have.

3. [Why are biomarker tests used in oncology?](#)

Biomarker tests can be used to:

- a. Determine whether you are at risk of developing cancer.
- b. Determine whether you have cancer.
- c. Determine how aggressive your disease is.
- d. Predict how your body will respond to a treatment.
- e. Determine how your disease might progress.

4. [How is biomarker testing performed?](#)

Biomarker testing involves collecting a sample of your cancer cells through surgery, tissue biopsy, or a liquid biopsy.

5. [What happens after I get tested?](#)

Your samples will be sent to a special lab where they will be tested for certain biomarkers. The lab will provide you with a list of the biomarkers in your cancer cells and potentially beneficial treatments.

6. [Do I have a right to biomarker testing?](#)

As a patient you have the right to engage in shared decision-making with your healthcare team when discussing your personalized treatment plan, which may include the use of biomarker testing.

7. [What questions should I ask my healthcare team before and after biomarker testing?](#)

Empower yourself with information by arriving at each appointment with a list of questions to ask your healthcare team.

8. [How to get additional resources and support](#)

9. [Glossary of common biomarker cancer testing terminology](#)

10. [Acknowledgment](#)

[Useful guidance for the adaptation of the resource](#)

Is biomarker testing part of precision medicine?¹

Yes, biomarker testing is an important part of precision medicine. Precision medicine is an approach to medical care in which disease prevention, diagnosis, and treatment are tailored to the genes, proteins, and other substances in your body.

For cancer treatment, precision medicine means using biomarkers and other tests to select treatments that are most likely to help you, while sparing you from receiving ineffective treatments.

The idea of precision medicine isn't new, but recent advances in science and technology have helped speed up the pace of this area of research. Scientists now understand that cancer cells can have many different changes in genes, proteins, and other substances that make the cells grow and spread. They have also learned that even two people with the same type of cancer may not have the same changes in their cancer. Some of these changes affect how certain cancer treatments work.

Even though researchers are making progress every day, the precision medicine approach to cancer treatment is not yet part of routine care for most patients. But it is important to note that even the "standard" approach to cancer treatment (selecting treatments based on the type of cancer you have, its size, and whether it has spread) is effective and is personalized to each patient.

What is biomarker testing for cancer treatment?



Instructions for adaptation:

Please read the text below, check that the information applies to your patient population and edit it with disease-specific information where needed. If there is any additional information that should be included for your patient population, include it in the relevant sections.

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 abbreviation. Examples of biomarkers are HER2 in breast cancer or EGFR in lung cancer.²

Biomarker testing can help to diagnose or identify the type of cancer you have. A biomarker can also be referred to as a marker, tumor marker, genomic marker, signature molecule, or molecular diagnostic³.

¹ National Cancer Institute (2021) Biomarker Testing for Cancer Treatment.

² Cancer Support Community (2018) Precision Medicine Plain Language Lexicon.

³ Lungevity. (2021) What you need to know about... biomarker testing.

Biomarker testing can be done to look for mutations in a cancer (called somatic mutations). Somatic mutations cause most cancers and can't be passed on to family members.⁴

This is different from genetic testing, which looks for mutations in your genes that are inherited from your parents and can be passed on to children⁵. The latter one is not covered in this resource.

Why are biomarkers tests used in oncology?



Instructions for adaptation:

Please read the text below, check that the information applies to your patient population and fill in the blank spaces with disease-specific information where needed.

If there is any additional information that should be included for your patient population, include it in the relevant sections.

Biomarker testing can help identify a specific mutation in a cancer cell, reveal the “drivers” that can cause cancer to grow in different ways, or predict how well your body responds to certain types of treatment. More specifically, these are the uses of biomarker testing in oncology. It can be used to determine:

- **Risk (also called susceptibility):** to help assess how likely you are to develop cancer or determine whether you should undergo more intensive screening.
- **Diagnosis:** to help detect or confirm a disease or condition⁶.
- **Prognosis:** to forecast how your cancer will progress in the absence of therapy.
- **Prediction:** to help assess whether you will respond to treatment.
- **Response:** to help predict your response — both beneficial or harmful — to a certain treatment⁷.
- **Monitoring:** to help determine how you are doing over time, either on or off treatment⁸.
- **Safety:** to help measure the likelihood, presence, or extent of side effects before or after undertaking a treatment⁹.

In the figure below, you can check how biomarker testing can be used in different moments of the patient pathway.

⁴ National Cancer Institute (2021) Biomarker Testing for Cancer Treatment

⁵ Cancer Support Community (2018) Precision Medicine Plain Language Lexicon.

⁶ FDA-NIH Working Group. BEST (Biomarkers, EndpointS, and other Tools) Glossary.

⁷ FDA-NIH Working Group. BEST (Biomarkers, EndpointS, and other Tools) Glossary.

⁸ Research Advocacy Network (2021) Precision Medicine in Oncology.

⁹ FDA-NIH Working Group. BEST (Biomarkers, EndpointS, and other Tools) Glossary.

USES OF BIOMARKERS IN CANCER CARE							
Before Diagnosis	Diagnosis	After Cancer Diagnosis					Post-Treatment
Risk	Diagnostic	Prognostic	Predicting treatment response	Response	Safety	Monitoring treatment response	Monitoring for recurrence
<i>Am I at increased risk of cancer?</i>	<i>Do I have cancer? What type of cancer do I have?</i>	<i>What is the expected course of my cancer?</i>	<i>Will my cancer respond to this treatment?</i>	<i>Should I receive a normal or lower dose or no dose?</i>	<i>What side effects am I likely to experience?</i>	<i>How is my cancer responding to this treatment?</i>	<i>Will my cancer come back?</i>

Fig. 1 Use of biomarkers in cancer care¹⁰. Adapted from Research Advocacy Network “Precision Medicine in Oncology”

Knowing your cancer’s biomarkers can **help you and your healthcare team make fully informed decisions** about how your cancer will be treated. **Ask your healthcare team about biomarker testing at any decision point** before new treatment begins, including upon diagnosis, recurrence, or progression¹¹.

Patients with a _____ cancer diagnosis should discuss biomarker testing with their healthcare team to understand what tests (e.g. single-gene testing, targeted panel testing, comprehensive genomic profiling, etc.) are recommended based on the type and stage of cancer and the most current clinical guidelines¹². Based on your _____ cancer diagnosis, it is recommended to discuss with your healthcare team to proceed with _____ testing.

Keep in mind that even when a cancer biomarker is present, there is no guarantee that a targeted therapy will work against your cancer. One reason for this is that the molecular makeup of cancer can change over time. This means that the target molecule of the biomarker can change. The tumor can also find ways to grow and spread without that molecule¹³. If a drug stops working or your cancer returns, that may mean that your tumor cells have become resistant to a treatment. At that point, another biomarker test may be needed to understand how your tumor has changed. This could identify new biomarkers that may be targeted for treatment¹⁴.

¹⁰ Research Advocacy Network (2021) Precision Medicine in Oncology.

¹¹ Lungevity (2021) Biomarker Conversation Starter.

¹² Lungevity (2021) Patients Bill of Rights.

¹³ Cancer Support Community (2018) Precision Medicine, Biomarker Testing.

¹⁴Cholangiocarcinoma Foundation (2020) For Cancer Patients, Biomarkers Matter.

How is biomarker testing performed?



Instructions for adaptation:

Please read the text below, check that the information applies to your patient population and fill in the blank spaces with disease-specific information where needed.

If there is any additional information that should be included for your patient population, include it in the relevant sections.

TISSUE BIOPSY

If you and your healthcare team decide to make biomarker testing part of your care, they will take a sample of your cancer cells.

Currently, for _____ [e.g. lung cancer], biomarker testing is performed as _____ [e.g., tissue biopsy] ¹⁵.

If you have, or are suspected to have, a solid tumor, they may take a sample during surgery. If you aren't having surgery, you may need to have a **tissue biopsy** of your tumor¹⁶. **Regardless of how the tissue is collected, a patient should confirm with the healthcare team, before the tissue is removed, that an adequate tissue sample will be collected so that all necessary biomarker tests can be performed.** Tissue samples from the tumor can be saved for a long time so that additional testing, if necessary, can be done¹⁷.

Among the tissue collection techniques are [e.g. bronchoscopy, transthoracic needle biopsy, etc...]

Biomarker testing can be done on both primary tumors and metastatic tumors. If the tumor sample is too small to run through multiple tests, priority should be given to testing for mutations that are the most likely to be present, have an _____ [e.g. FDA or EMA]-approved drug treatment, or otherwise help with treatment decisions. Therefore, at this time, if there is only a limited amount of tumor tissue, _____ cancer should be tested for the _____ mutations^{18,19}.

¹⁵ There are several international guidelines that can help you add the cancer-specific biomarkers; review the section with [useful resources](#). We would advise you to check national guidances too.

¹⁶ National Cancer Institute (2021) Biomarker Testing for Cancer Treatment.

¹⁷ Lungevity (2021) What you need to know about... biomarker testing.

¹⁸ There are several international guidelines that can help you add the cancer-specific biomarkers; review the section with [useful resources](#). We would advise you to check national guidances too.

¹⁹ Lungevity (2021) What you need to know about... biomarker testing.

Consider also that biomarker testing can help you and your healthcare team identify appropriate clinical trials.

LIQUID BIOPSY

If you are getting a biomarker test known as a **liquid biopsy**, you will need to have a blood draw. You might get a liquid biopsy test if you can't safely get a tumor biopsy, for example, because your tumor is hard to reach with a needle²⁰ or if you have a health condition that prevents you from undergoing the procedure. For some cancer types, a liquid biopsy can detect cancer cells of a tumor circulating in the blood or pieces of the tumor's DNA circulating in your blood. This test may also be used as a less invasive method of monitoring progression or to assess whether the cancer has become resistant to the treatment. Keep in mind, however, that not all cancers shed cells into the bloodstream in the same way. Therefore, a liquid biopsy may only be useful for detecting or monitoring certain cancers. In many cases, tissue biopsies may be more accurate than liquid biopsies.

Finally, for biomarker tests that analyze genes, you will also need to give a sample of your healthy cells. This is usually done by collecting your blood, saliva, or a small piece of your skin. These tests compare your cancer cells with your healthy cells to find genetic changes (called somatic mutations) that arose during your lifetime. Somatic mutations cause most cancers and can't be passed on to family members²¹.

What happens after I get tested?

Your samples will be sent to a lab where they will be tested for certain biomarkers. The lab will create a report that will show if any biomarkers were identified in your cancer cells that may be helpful in selecting the right treatment approach for you²².

The test results can approximately take up to _____ weeks to be received by your healthcare team.

It is a good idea to get a copy of your biomarker testing report for your own information and to have it available to show to other healthcare providers, if necessary.

²⁰ National Cancer Institute (2021) Biomarker Testing for Cancer Treatment.

²¹ National Cancer Institute (2021) Biomarker Testing for Cancer Treatment.

²² National Cancer Institute (2021) Biomarker Testing for Cancer Treatment.

Do I have a right to biomarker testing ²³?



Instructions for adaptation:

Please read the text below, check that the information applies to your patient population and fill in the blank spaces with disease-specific information where needed. If there is any additional information that should be included for your patient population, include it in the relevant sections.

As a patient, you have the right to:

- **Receive optimal care and engage in shared decision-making** with your healthcare team when discussing your personalized treatment plan.
- **Understand the cost of comprehensive biomarker testing** and whether it will be covered by _____
- **Know whether your healthcare team has already completed biomarker testing** on your _____ cancer, and if so, what the results of your test show.
- **Access and understand your biomarker test results**, which has critical information about whether you may respond to _____ treatment.
- **Request a print or electronic copy of your biomarker testing report** for your medical records, which can be helpful for seeking a second opinion.
- **Seek a second opinion** about your treatment options based on your biomarker test results.

What questions should I ask my healthcare team before and after biomarker testing?



*The questions below are part of a broader **Q&A Resource Builder**. This resource allows you to have a comprehensive list of precision medicine questions that patients can ask their HCPs. Use the list of questions below and adapt them as needed.*

UNDERSTANDING DIAGNOSTIC PROCEDURES

- What diagnostic tests do you recommend for my condition and why? What can the test results tell us about my condition?
- What will the tests involve? Are they available to me? How much time/energy will it take of me?

²³ Lungevity (2021) Patients Bill of Rights.

- When and where will I have my testing completed? Will all testing be completed now or will some be done later?
- Is there a waiting period to have any tests done? How long will it take to get the results?
- How will I be given my results? Who can help me understand them? Can I have a copy of my test results?
- What is the difference between genetic and biomarker (mutation, genomic, or molecular) testing?
- Do you recommend biomarker (mutation, genomic, or molecular testing) testing?
 - If so, which biomarkers are important for this particular type of cancer?
 - If so, which testing method is most appropriate for this type of cancer and why?
 - If so, what types of treatments might biomarker testing identify? If my biomarker test results show I am a candidate for precision medicine, what would the next steps be?
- Might there be a genetic (hereditary) explanation for my condition? Do you recommend genetic testing? Should other members of my family get tested?
- Is there counseling available for me to discuss the outcome?
- May I need to pay towards my tests? If so, are there financial assistance programs or resources you can recommend? (in countries where relevant)

UNDERSTANDING TREATMENT OPTIONS

- Which treatment or combination of treatments would you recommend for me and why?
- Are there other treatment options available, such as clinical trials? How can I access them and what do they involve?
- What does each treatment aim to do and how effective is it likely to be? Is there anything I can do myself to help?
- Do I need to have further tests before initiating the treatment? Will we wait to get all of the test results back before starting treatment?
- When will the treatment start? Is there time to wait and see, and time for me to consider my options?
- What are the potential side effects of the recommended treatments? How might they affect my quality of life? And what can we do to manage them?
- Will the recommended treatment interact with other medications I am taking for my condition or vice versa? How can we manage this?
- How do I take my treatment and how often? How long will it last?
- I would like to get a second opinion before I commit to my treatment plan. Can you suggest a suitable specialist?
- Is there any psychological/social/emotional support or tools available during my treatment to support me and/or my family members?
- May I need to pay towards my tests? If so, are there financial assistance programs or resources you can recommend? (in countries where relevant)

How to get additional resources and support

We would advise using this section to signpost to your organization's website, helpline, support groups, patient stories, or other resources that might be helpful for patients.

Glossary of common biomarker cancer testing terminology

- **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 abbreviation. Examples of biomarkers are HER2 in breast cancer or EGFR in lung cancer (Cancer Support Community, Precision Medicine Plain Language Lexicon).
- **Biomarker testing** (also referred to as *Comprehensive biomarker testing, Gene-based cancer testing, Genetic testing of the cancer, Cancer marker testing, Next generation sequencing, Genomic profiling, Mutation biomarker testing, Genomic testing, Molecular profiling, Tumor marker testing, Mutation testing, Molecular 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 the cancer in your body (Cancer Support Community, Precision Medicine Plain Language Lexicon).
- **Diagnostic test:** a type of test used to help diagnose a disease or condition (National Cancer Institute, Dictionary of Cancer Terms).
- **Genetic testing:** 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 find out how well treatment is working (National Cancer Institute, Dictionary of Cancer Terms).
- **Liquid biopsy:** (also referred to as *fluid 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. If test results are negative, your doctor may want to do a tissue biopsy (Cancer Support Community, Precision Medicine Plain Language Lexicon).

- **Secondary tumor:** *(also referred to as metastatic tumor)* a term used to indicate the spread of cancer from the primary site (place where it started) to other places in the body (National Cancer Institute, Dictionary of Cancer Terms).
- **Mutation in the cancer:** Two kinds of mutations can be found in cancer cells — mutations you inherit and mutations you acquire during your lifetime (called somatic mutations). Inherited mutations are found in all your cells. Acquired mutations may only be found in the cancer's cells. Acquired mutations happen as you get older and are the result of 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 (Cancer Support Community, Precision Medicine Plain Language Lexicon).
- **Precision medicine:** a form of medicine that uses information about a person's own 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 make a prognosis (National Cancer Institute, Dictionary of Cancer Terms).
- **Primary tumor:** a term used to describe the original, or first, tumor in the body. Cancer cells from a primary tumor may spread to other parts of the body and form new, or secondary, tumors. This is called metastasis. These secondary tumors are the same type of cancer as the primary tumor. Also called primary cancer (National Cancer Institute, Dictionary of Cancer Terms).
- **Side effect:** *(also referred to as adverse event)* a problem that occurs when treatment affects healthy tissues or organs. Some common side effects of cancer treatment are nausea, vomiting, fatigue, pain, decreased blood cell counts, hair loss, and mouth sores (National Cancer Institute, Dictionary of Cancer Terms).
- **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 (Cancer Support Community, Precision Medicine Plain Language Lexicon).

Acknowledgments

This resource was co-created by a global, multi-stakeholder working group of the [From Testing to Targeted Treatments](#) (FT3) Program.

FT3 brings together diverse stakeholders and activities in precision medicine to identify shared needs, build on existing good practices and learnings and make them replicable through practical and actionable tools and resources for precision medicine champions to drive real change and impact, starting with cancer and testing.

Ultimately, the goal is to develop integrated and optimized resources for different stakeholders to improve access to precision medicine, and to develop a positive momentum to be able to improve patient outcomes and experiences. This was developed thanks to the FT3 Members and Community of Practice.

This guide is the result of the aggregation of resources originally developed by LUNGeVity, Lung Cancer Europe, the National Cancer Institute, Research Advocacy Network, Cancer Support Community, and others, which emerged as good resources in terms of scientific credibility, patient centricity, plain language, and ease of use. We thank them for their good practice inspiration.

The following resources were consulted in developing this adaptable resource:

- Cancer Support Community (2018) Precision Medicine: Biomarker Testing. Available at: https://www.youtube.com/watch?v=Pciz_jZYGOU
- Cholangiocarcinoma Foundation (2020) For Cancer Patients, Biomarkers Matter. Available at: <https://www.youtube.com/watch?v=Hwo-kxBOoww>
- European Cancer Patient Coalition (2018) Cancer Biomarkers in the Era of Personalised Medicines. Available at: https://www.youtube.com/watch?v=t_7wuuZAOAg&t=77s
- Lung Cancer Europe. Factsheet: What is Biomarker Testing? Available at: <https://www.lungcancereurope.eu/wp-content/uploads/2021/07/LuCE-Factsheet-Biomarker-testing.pdf?fbclid=IwAR3yuXlIei5lpqIjwHNrvRcOp3ouECInkprAtcozN-2fH1q4zxCvtfPsK4g>
- Lungevity (2021) Comprehensive Biomarker Testing: Why It's Important for Lung Cancer. Available at: <https://www.lungevity.org/sites/default/files/nom/NOM-Biomarker-Quick-Facts-041421.pdf>
- Lungevity (2021) Patients Bill of Rights. Available at: <https://www.lungevity.org/sites/default/files/nom/NOM-Patients-Bill-of-Rights-041421.pdf>
- Lungevity (2021) Biomarker Conversation Starter. Available at: <https://www.lungevity.org/sites/default/files/nom/NOM-Biomarker-Conversation-Starte-041421r.pdf>
- Lungevity. (2021) What you need to know about biomarker testing. Available at: <https://www.lungevity.org/sites/default/files/request-materials/biomarker-testing-booklet-081121.pdf>
- Lungevity. Biomarker Testing. Available at: <https://www.lungevity.org/for-patients-caregivers/navigating-your-diagnosis/biomarker-testing>

- National Cancer Institute (2021) Biomarker Testing for Cancer Treatment. Available at: <https://www.cancer.gov/about-cancer/treatment/types/biomarker-testing-cancer-treatment>
- Research Advocacy Network (2021) Precision Medicine in Oncology. Available at: https://researchadvocacy.org/advocate-institute/precision-medicine-oncology-educational-resources?_ga=2.166125884.328009269.1641898522-1479582868.1636559867

Useful guidance for the adaptation of this resource

When adapting your resource you can make use of the following materials that can help you find the information you need.

Guidelines:

- American Society of Clinical Oncology (ASCO). Guidelines by Clinical Area, Available at: <https://www.asco.org/practice-patients/guidelines>. These clinical practice guidelines provide recommendations to serve as a guide for doctors and outline appropriate methods of treatment and care.
- European Society for Medical Oncology (ESMO). Cancer Guides for Patients. Available at: <https://www.esmo.org/for-patients/patient-guides>. These guides translate complex medical information into a language understandable to patients. The medical information is based on the ESMO Clinical Practice Guidelines.
- National Comprehensive Cancer Network (NCCN). Guidelines for Patients. Available at: <https://www.nccn.org/patientresources/patient-resources/guidelines-for-patients>. This library of over 62 guidelines for patients with cancer offers plain-language recommendations on diagnostic procedures and treatment options from the NCCN Guidelines.

Glossaries:

- Cancer Support Community. Precision Medicine Plain Language Lexicon. Available at: https://www.cancersupportcommunity.org/sites/default/files/fsac/Precision_Medicine_Lexicon_w_Introduction-Nov_2021-Updated_with_survey_results.pdf
- Lungevity. Glossary. Available at: <https://www.lungevity.org/for-patients-caregivers/helpful-resources/glossary>
- National Cancer Institute. Dictionary of Cancer Terms. Available at: <https://www.cancer.gov/publications/dictionaries/cancer-terms/expand/B>
- National Cancer Institute. NCI Dictionary of Genetics Terms. Available at: <https://www.cancer.gov/publications/dictionaries/genetics-dictionary/expand/B>
- National Human Genome Institute. Talking Glossary of Genetic Terms. Available at: <https://www.genome.gov/genetics-glossary/s#glossary>
- Oncology Nursing Society. Genomics Taxonomy. Available at: <https://www.ons.org/genomics-taxonomy>
- U.S. Food and Drug Administration and the National Institutes of Health. BEST (Biomarkers, EndpointS, and other Tools) Glossary. Available at: <https://www.ncbi.nlm.nih.gov/books/NBK338448/>

