



# The Patient Guide On Cancer Biomarkers



European Cancer  
Patient Coalition

Produced in collaboration with

**LEK**<sup>TM</sup>

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European Cancer  
Patient Coalition

The Patient Guide  
On Cancer Biomarkers

# Table of Contents

## A. General Information

<b>1. What are cancer biomarkers?</b>	<b>5</b>
<b>2. What is the relation of precision oncology to cancer biomarker testing?</b>	<b>5</b>
· What is precision oncology?	
· Cancer biomarkers and precision oncology	
· How a patient knows that he/she is receiving precision oncology treatment	
<b>3. Is cancer biomarker testing available in all EU member states and the UK?</b>	<b>8</b>
· Summary of results of a survey about availability of cancer biomarkers in Europe	
· What are the barriers to accessing cancer biomarker testing?	
· How can these barriers be overcome?	
· What patients and patient organizations can do	

## B. Biomarker testing in cancer

<b>4. Why should I have biomarker testing?</b>	<b>11</b>
<b>5. How is cancer biomarker testing performed?</b>	<b>11</b>
<b>6. Where can I have cancer biomarker testing performed?</b>	<b>12</b>
<b>7. Can I ask for cancer biomarker testing to be performed?</b>	<b>12</b>
· To which doctor should the request be directed?	
<b>8. For which cancer types are biomarkers available?</b>	<b>12</b>
· Is there one or more biomarkers per cancer type?	
· Are these biomarkers available in all cancer hospitals?	
· If they are not available in the hospital in which I am treated, what can I do?	
<b>9. My doctor tells me that he/she cannot prescribe the cancer biomarker testing that I need ....</b>	<b>12</b>
· Why is the cancer biomarker test that I require not reimbursed?	
· What can the doctor do?	
· How can reimbursement or funding for my tests be secured?	
<b>10. If the test suitable for me is not available, what can I do as a patient?</b>	<b>13</b>

## C. Information for patients and carers

<b>11. Get prepared for your medical appointment with information and with a list of questions to ask your doctor</b>	<b>14</b>
<b>12. What questions should I ask my healthcare team before and after cancer biomarker testing?</b>	<b>15</b>
<b>13. How to get additional resources and support</b>	<b>16</b>
<b>14. Glossary of common cancer biomarker testing terminology</b>	<b>17</b>
<b>15. References</b>	<b>18</b>



## Foreword

The burden of cancer continues to grow globally, creating substantial pressure on patients, their families, and communities. Cancer represents the second largest cause of death and morbidity in Europe, with more than 3.7 million new cases and 1.9 million deaths each year [1].

However, new therapeutic approaches are bringing us closer to a future where cancer becomes a curable disease. Knowledge of cancer has improved vastly in the last two decades, revealing the huge variability not only between cancer types (e.g. breast cancer, colorectal cancer), but also between patients with the same cancer type (e.g. triple negative breast cancer, erbB2 positive breast cancer), and highlighting the need for – and the promise of – tailoring cancer care to individual patient characteristics.

Fuelled by this knowledge, cancer treatment is increasingly shifting towards precision oncology, an approach that systematically utilises patient/tumour data to inform personalised treatment decisions. With precision oncology, physicians can identify molecular characteristics of a patient's tumour and select a treatment that will be most effective at targeting that specific tumour subtype. The vision for precision oncology is transformative: to deliver superior outcomes for all cancer patients and ultimately reduce the suffering caused by cancer.

The Cancer Biomarkers Patient Guide was initiated by the European Cancer Patient Coalition – ECPC as part of ECPC activities for raising awareness on personalised medicine in cancer to assist patients, their family members and friends to better understand the cancer biomarker testing that may influence the decisions for their personalised medicine treatment. The Guide has been prepared to offer concise information to patients, who will have cancer biomarker testing as part of receiving personalized medicine treatment. The Guide includes all basic information cancer patients, carers and families may wish to have at hand in preparation of their medical appointments.

Cancer patients are invited to consult the Glossary to familiarize themselves with the terms used for cancer biomarker testing as well as the suggested questions to be asked to their healthcare team before and after cancer biomarker testing.

We hope that this Guide will be useful to you and we invite you to share any suggestions you may have for its improvement with us, by mail at [info@ecpc.org](mailto:info@ecpc.org) – [kathi.apostolidis@ecpc.org](mailto:kathi.apostolidis@ecpc.org).

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## A. General Information

### 1. What are cancer biomarkers?

#### Cancer Biomarker

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. Processes that a biomarker measures could include cell death, cell growth, resistance to chemotherapy, etc. "Biomarker" refers to any molecule in the human body that can be measured to assess health (e.g., mutation in one of the BRCA genes as an indicator of breast or ovarian cancer risk). Cancer biomarkers can be detected from blood, other bodily fluids or tissue [3, 4]



#### Biomarker test

A **biomarker test** is a measurement developed to detect and/or quantitate one, or several, biomarkers for the screening, diagnosis and/or prognosis of cancer patients. It includes tests looking for the presence of abnormal proteins and tests looking for abnormal changes within the genetic/genomic material of the patient [3, 4]

#### Companion diagnostic / companion biomarkers

A Companion Diagnostic is a test that is linked to the use of a particular treatment, to identify patients who are most likely to benefit from the treatment, those who will receive no benefit from the treatment or those who may be at increased risk of serious side effects because of the treatment

### 2. What is the relation of precision oncology to cancer biomarker testing?

#### Precision oncology

**Precision oncology** is a healthcare approach that utilises molecular information and health data from an individual patient to generate care insights to prevent or treat cancer in a manner tailored to that specific patient's tumour subtype [5]. Precision oncology has the potential to transform health outcomes for patients.



## Benefits of precision oncology [2]:

Benefits	
<b>Enables more targeted treatment</b>	Precision oncology and the deployment of cancer biomarkers can be used to predict if a treatment will work well in particular patients, and if so, deliver that treatment to the patients selected based on the result of their cancer biomarker test. Given that it is a more targeted approach than the one size fits all chemotherapy, it has the potential to be more effective than a traditional therapy
<b>Limits the use of ineffective drugs</b>	Treatments are tailored specifically to the characteristics of a patient's cancer, so there is less chance of being treated with medicines that have lower efficacy
<b>Causes fewer side effects</b>	Precision medicines target the specific proteins, genes or processes that are driving a patient's cancer, so can have a reduced impact on healthy cells in the body compared with traditional chemotherapy, so they can have a reduced impact
<b>Lowers the treatment burden</b>	Precision oncology increases the likelihood of finding the most suitable treatment at the first treatment intervention, thus reducing the need for patients to take several medications before the correct one is identified. This can also reduce the length of hospital stays

Biomarkers are essential tools in the diagnosis and treatment of cancer because they can be used to provide precise diagnoses and identify whether a patient will or will not respond to treatment, therefore informing treatment selection. They can also help predict and monitor disease progression and identify whether a patient is at increased risk of developing a given condition. Biomarker testing for precision oncology can be separated into two broad types:

### Single biomarker testing

*Single biomarker testing* evaluates the presence of a single gene mutation or expression of a single protein within a biopsy (or blood) sample associated with a particular form of cancer (e.g., HER2 testing in breast cancer patients).



### Comprehensive multi-biomarker testing

*Comprehensive biomarker testing* (e.g., next-generation sequencing (NGS)) refers to the evaluation of tumour or blood samples to detect multiple changes to genes that are known to be involved in the patient's cancer.

The shift towards precision medicine is supported by significant advances in biomarker testing, with NGS allowing the detection of genomic alterations which drive tumour development and providing critical insights into patients' likely response to treatment and the progression of their disease.

## Pros and cons of Next Generation Sequencing (NGS) [6]:

Pros	Cons
NGS allows the testing of many genes simultaneously, negating the need for multiple tests, which may be particularly useful when limited biopsy tissue is available	Results of NGS testing typically take two to three weeks to arrive, compared with a week or less for single biomarker tests
Testing several genes simultaneously with NGS can reduce the time between testing and initiation of treatment, compared with performing several single gene tests one after the other	The cost of NGS is typically significantly higher than that of single biomarker testing (assuming additional single biomarker tests are required – if additional single gene tests are required, then NGS can become more cost effective)
NGS can identify new cancer biomarkers beyond those commonly employed, potentially identifying additional treatment options	NGS can generate complex results that may be difficult to interpret, so may not lead to effective treatment decisions. This emphasises the need for education in cancer biomarkers and precision oncology, both for cancer patients and cancer healthcare professionals
The broad collection of cancer biomarkers identified via NGS may help identify patterns of patient responses and thus aid the refinement of precision oncology treatments	

In the context of precision oncology, you may also have cancer biomarker testing referred to as [7]:

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

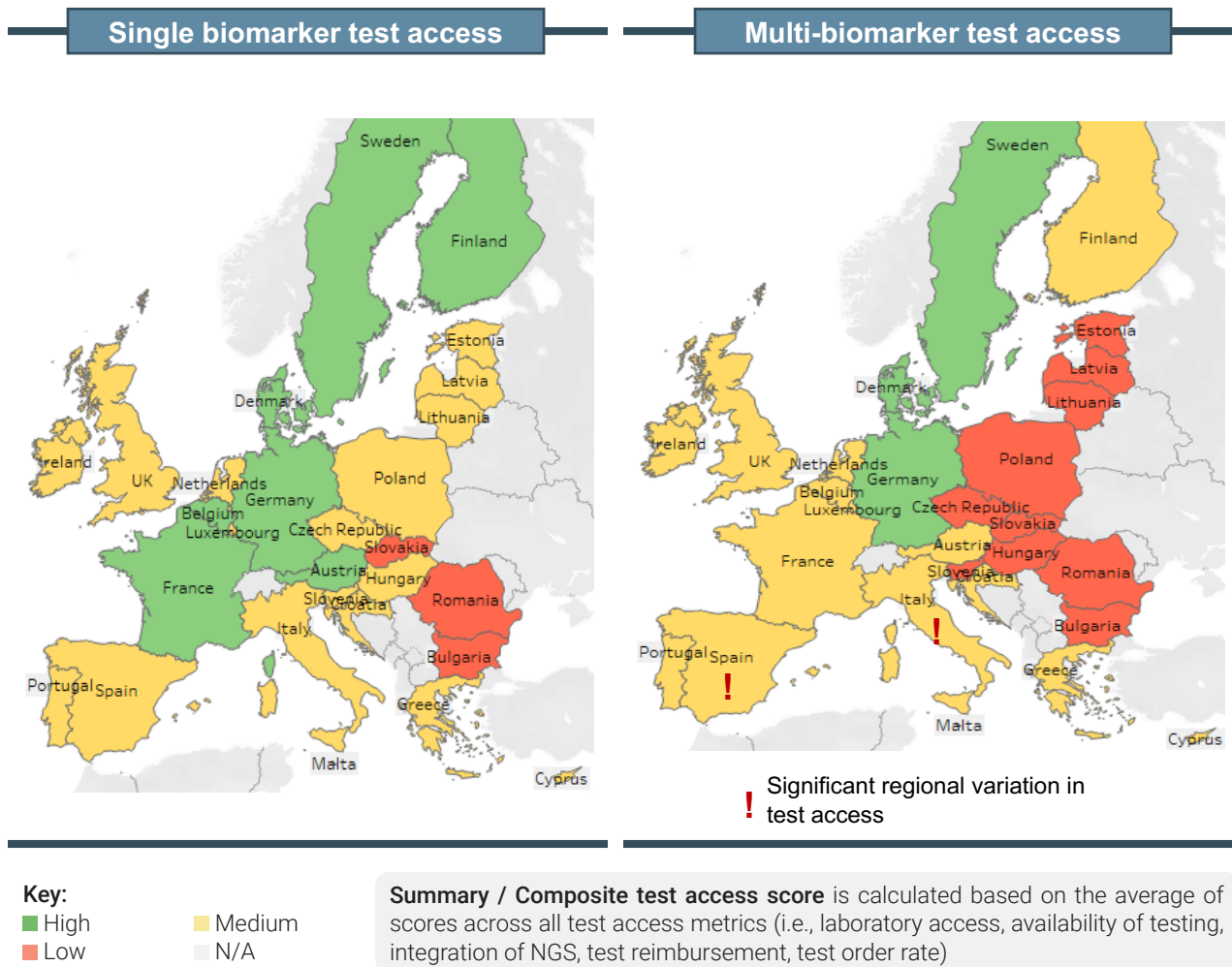




### 3. Is cancer biomarker testing available in all EU member states and the UK?

The promise of precision oncology can only be realized if patients across the whole of Europe have equitable access to the cancer biomarker testing required to determine if they are eligible for a particular treatment. If biomarker testing is not available, patients may not be able to benefit from the more effective targeted treatments for their specific cancer subtype, and instead might be limited to traditional treatment options that may be less effective.

#### Single vs. multi-biomarker test access in oncology by country, EU27 and UK [8]



*Single biomarker test access was analysed based on laboratory access, availability of single biomarker testing, single biomarker test reimbursement and single biomarker test order rate.<sup>1</sup>*

The analysis found that there are still gaps in access to single biomarker testing in oncology in some European countries, e.g., due to a lack of laboratory infrastructure, limited funding of tests or delays in getting access to tests.

<sup>1</sup> Test access was defined based on a country's performance on the following criteria: how many diagnostic labs exist which can perform biomarker testing; to what extent the biomarker test cost is paid for by the public healthcare system; and the share of eligible patients who receive biomarker tests



Currently, access to biomarker testing is not equal across European countries (EU27 and UK) [8]:



- In 15 out of 28 European countries, it takes one year or more from the reimbursed launch of a precision medicine until the corresponding single biomarker test becomes available (these are: the UK, Ireland, Luxembourg, Austria, Denmark, Sweden, Estonia, Greece, Malta, Cyprus, Slovenia, Slovakia, Romania, Czech Republic and Bulgaria)
- In 13 out of 28 European countries, single biomarker testing is performed in fewer than 75% of biopsies from patients who are theoretically eligible for the test

*Multi-biomarker test access was analysed based on laboratory access, availability of NGS testing, integration of NGS into clinical practice<sup>2</sup>, NGS test reimbursement, and NGS test order rate.*

Access to NGS-based multi-biomarker testing is generally lower than single-biomarker testing as it is a newer, more complex, and more expensive technology. Availability is increasing, but it remains limited in many European countries, particularly in Eastern and Southern Europe, and NGS is often not accessible for all types of cancer.

In summary, there is significant regional disparity in biomarker test access across the EU27 and the UK: Northern and Western European countries generally perform the highest on the metrics mentioned above, reflecting their higher investment in healthcare.

A number of barriers to accessing cancer biomarker testing exist across Europe [2]:

## Barriers



Limited availability of precision medicines linked to cancer biomarkers

Unclear approaches for assessing the value of diagnostic tests to healthcare systems and to patients

Insufficient laboratory infrastructure, test capabilities and referral pathways in different European countries

Limited availability of public funding to support cancer biomarker testing

Limited stakeholder awareness and education of the availability and benefits of cancer biomarker testing

<sup>2</sup> Defined as time to introduction of any NGS test modality in the country, plus NGS uptake: average % of all biopsies currently analysed using NGS technology

**Without immediate and concerted action to ensure the provision of adequate biomarker testing across countries, it is impossible to harness the full benefits of precision oncology.**



The European Federation of Pharmaceutical Industries and Associations (EFPIA), the International Quality Network for Pathology (IQN Path) and the European Cancer Patient Coalition (ECPC) have developed recommendations to encourage policymakers to improve access to biomarker testing for patients across Europe [2]. However, it is important that patients and patient organisations also take action to overcome barriers to accessing biomarker testing. Patient advocacy groups can play a key role by developing and distributing information on the purpose and benefits of biomarker testing and the specific test options available. In a recent cancer-focused survey, patients in 15 out of 16 surveyed countries rated their satisfaction with information received about the biomarker testing procedure, test results and implications for treatment as low to medium [9]. Ultimately, patients who are more knowledgeable about cancer biomarker testing and precision oncology treatment options can, and should, actively demand these approaches to be employed in the treatment of their cancer.

### **Potential actions for patients:**

#### **Actions**



Seek out materials published by cancer patient organisation on the benefits of cancer biomarker testing and its role in precision oncology

Keep up to date with any changes in cancer biomarker testing guidelines or the availability of new cancer biomarker tests, via your cancer patient organisation or by talking to your physician

Be aware of any regional cancer biomarker testing centres in your country so that you can inform your physician (e.g., national networks for biomarker testing in Germany, Sweden, the Netherlands and England; example: Genomic Laboratory Hubs in England)

Stay up to date with potential funding sources for cancer biomarker testing in your country, via information provided by your cancer patient organisation, so that you can actively pursue reimbursement options

Allow your data to be used for cancer research, to accelerate the development of precision oncology

Actively encourage your physicians to consider cancer biomarker testing and associated precision oncology treatments at the earliest possible opportunity

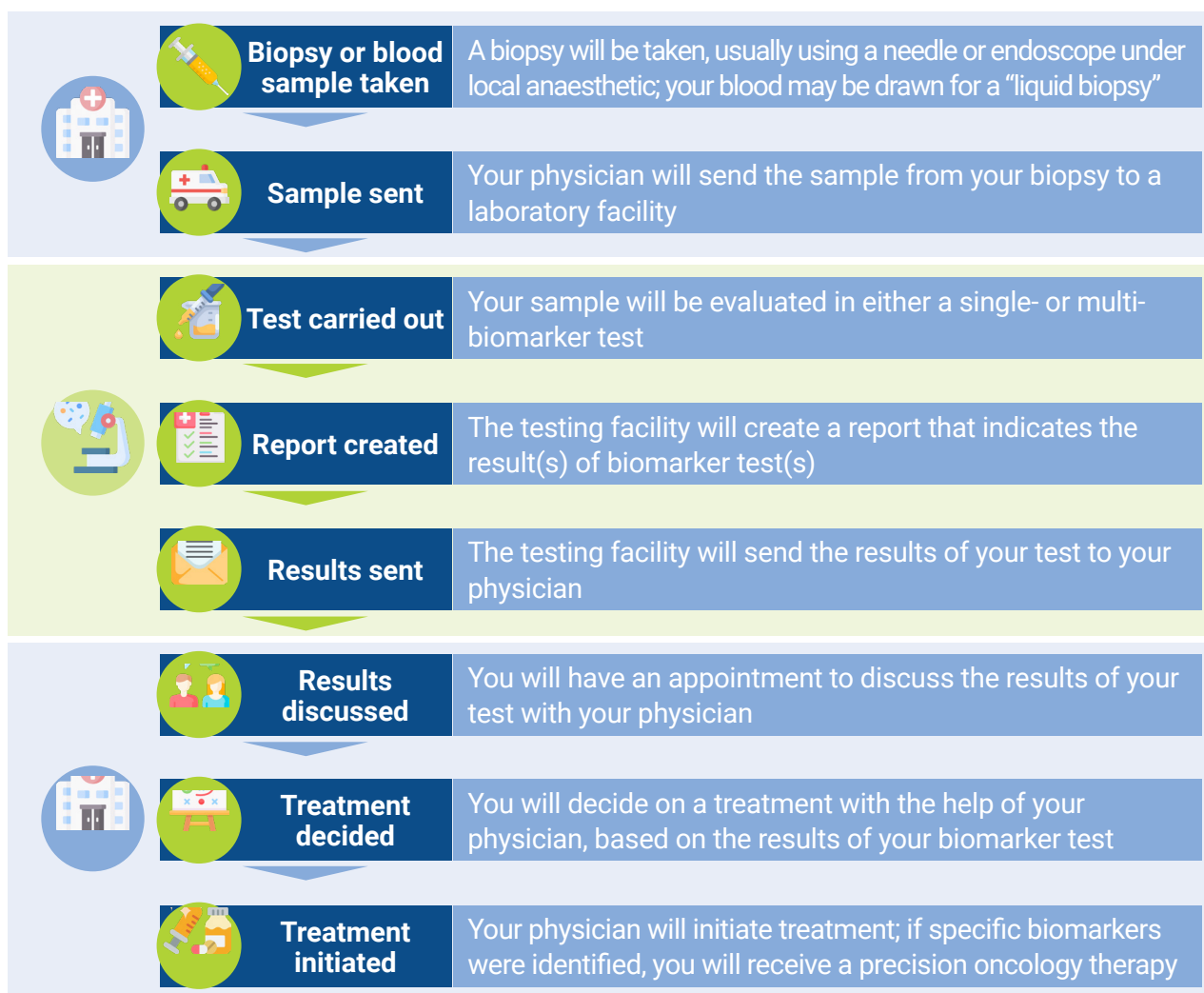
## B. Biomarker testing in cancer

### 4. Why shall I have cancer biomarker testing?

Cancer biomarker testing can help you and your physician select the most appropriate treatment for your cancer. Precision oncology therapies may only work for patients that have specific biomarkers associated with specific cancer subtypes. Testing can therefore identify which treatment should be provided that is most likely to be effective for your cancer. For example, patients with cancers caused by mutations in a gene known as the Epidermal Growth Factor Receptor (EGFR) can be treated effectively with drugs that stop this EGFR protein from working in the cancer cell (EGFR inhibitors). Biomarker testing can be used to identify whether these changes in the EGFR gene are present, so that EGFR inhibitors can be prescribed only to those patients that will benefit from them.

### 5. How is cancer biomarker testing is performed?

Cancer biomarker testing can be used to detect specific biomarkers in tumour tissue or in the blood. The testing process is typically as follows [7, 10]:



## 6. Where can I have cancer biomarker testing performed?

Depending on availability of testing facilities, cancer biomarker testing may be performed at the hospital at which you are normally treated or sent by your hospital to regional or national laboratories. Alternatively, you may have to travel to regional diagnostic centres or specialised laboratories to receive a test.

## 7. Can I ask for cancer biomarker testing to be performed?

In some circumstances, physicians might not be aware of the possibility of cancer biomarker testing and precision oncology treatment options and hence not prescribe the test/treatment. As a patient, you should be prepared to ask your physician whether cancer biomarker testing is available and whether it could help you to receive the optimal treatment for your cancer. The oncologist or other physician who manages your treatment is the best person to ask, as they would be most likely to manage your referral to a cancer biomarker testing pathway and a precision oncology treatment if this is the right option for you.

## 8. For which cancer types are biomarkers available?

Cancer patients may discuss with their physicians the tests that are indicated for their condition. In most European countries, more than one biomarker test is available and reimbursed per cancer type. As a general indication, for the following common cancers, examples of the most widely used cancer biomarkers are the following (*examples only*):

- Breast cancer: estrogen receptor (ER), progesterone receptor (PR), human epidermal growth factor receptor 2 (HER2), BReast CAncer gene 1 and BReast CAncer gene 2 (BRCA1 / BRCA2)
- Non-small cell lung cancer: Epidermal Growth Factor Receptor (EGFR), Anaplastic Lymphoma Kinase (ALK), Proto-Oncogene 1, Receptor Tyrosine Kinase (ROS1), Proto-oncogene B-Raf (BRAF), Programmed Death-Ligand 1 (PD-L1)
- Prostate: Prostate-specific antigen (PSA), BReast CAncer gene 2 (BRCA2), Alpha-methylacyl-CoA racemase (AMACR), Prostate-Specific Membrane Antigen (PSMA)

However, availability and reimbursement of biomarker tests can vary by country or region, so it is important to discuss access to testing with the treating physician who can also advise on how to receive a given test if it is not available in your hospital.

## 9. My doctor tells me that he/she cannot prescribe the biomarker testing I need

In some countries, especially in Southern and Eastern Europe, not all cancer biomarker tests are available or publicly reimbursed. For example, a survey conducted in 2020 found that NTRK testing was not available in Poland. At the time of the survey, tests for PD-L1, EGFR and BRCA were not covered by public funding in Romania [2]. There may be many reasons why a specific biomarker test is not reimbursed in your country.



In these circumstances, you should ask your doctor or cancer patient organisation about alternative funding sources. For example, some pharmaceutical manufacturers may provide funding for specific cancer biomarker tests which are needed for the prescription of a precision oncology medicine. Whilst a lack of funding can provide a significant barrier to test access, you should ask your doctor to consider all options, and to advocate for you in any discussions they may have on securing access to testing.

Potential reasons why a cancer biomarker test is not reimbursed:

- Cancer biomarker tests, especially those performed via next-generation sequencing (NGS), can be expensive, and some countries don't yet have effective means for assessing their value
- There can be a delay in the reimbursement of a biomarker test after its corresponding precision medicine has been approved for reimbursement. A survey conducted in 2020 found a delay of 1 year or more until the biomarker test became available through public funding in 15 European countries [2]
- In some countries, regional variations exist, whereby a biomarker test is publicly reimbursed in one region but not in another



## 10. If the test suitable for me is not available, what can I do as a patient?

In the event that biomarker testing is not available, you may contact your cancer patient organization to explore if they can assist you. It may be possible to appeal directly to your insurer to request reimbursement for your biomarker test. Information about your insurer's appeals policy can typically be found in your insurance policy paperwork or website or you may directly ask your insurer.

An additional option may be to consider engaging with your relevant health technology assessment (HTA) agency, which is the body that makes decisions on which products (e.g., biomarker tests) should be reimbursed. However, it must be noted that the receptiveness of HTA agencies to patient input is variable by country, with some countries, e.g., England, permitting continued input from patients and patient advocacy groups, and others, e.g., France, having limited patient involvement beyond the two patient advocacy group representatives on the committee [12]. In countries where patients can provide input (e.g., England, Germany, Netherlands, Sweden, Poland [20]), you could consider making a request through your patient organisation for involvement in the HTA assessment. Participation could include activities such as written submissions to form part of the evidence base, oral submissions (i.e., personal testimony) or attending meetings (e.g., report review processes) [21]. For those wishing to learn more about the HTA process, a good source of information is the HTAi the Health Technology Assessment International website <https://htai.org/patient-and-citizen-involvement/>.



<b>Who is the HTAi?</b>	<p>HTAi is a global health technology assessment society, which provides an open platform for global collaboration that leverages collective intelligence to improve health outcomes worldwide</p> <p>Its mission is to promote the development, communication, understanding, and use of health technology assessments (HTAs) around the world</p> <p>Members include multidisciplinary advisors, academics, scientists, professionals, public and private organizations, students, patients and citizens</p>
<b>What does the HTAi do?</b>	<p>Drives global HTA advocacy through <i>Policy Forums, Annual Meeting, and HTAi Interest Groups</i></p>
<b>How does the HTAi facilitate patient involvement with HTA?</b>	<p>The <i>HTAi Interest Groups (IGs)</i> enable patients and citizens to get involved and collaborate with staff from HTA bodies, government, research, industry (health technology developers). The <i>IGs</i> aim to strengthen HTA with the systematic incorporation of patient and citizen perspectives</p>
<b>How does a patient apply?</b>	<p>IGs are open to all members of HTAi. To join, or if you have further questions, you can contact: <a href="mailto:interestgroups@htai.org">interestgroups@htai.org</a></p>

## C. Information for patients and carers

### 11. Get prepared for your medical appointment with information and with a list of questions to ask your doctor

It is important to prepare yourself for your appointment with your physician, so that you are well-informed about your cancer and can ask the right questions about your cancer biomarker testing options. You should familiarise yourself with materials published by cancer patient organisations on cancer biomarker testing, including updated guidelines and test availability, the locations of your nearest regional testing centres, and the availability of funding to cover the costs of tests. For example, the German patient organisation Deutsche Krebshilfe publishes a series of guides on treatment guidelines, treatment and testing centres [19]. It should be noted that cancer patient organizations focus on specific areas of cancer support and your local cancer patient organization may not have information about biomarker testing, but you may ask if they can guide you to the relevant information sources.

If you have not yet undertaken your cancer biomarker test, you should prepare a list of key questions that you want to ask your physician, using the 'Questions to ask **before** biomarker testing' below as a guide. You should aim to come out of your appointment feeling comfortable that your physician has recommended the cancer biomarker test that is best for you, and that it can help to guide your treatment.

Equally, when you have had your cancer biomarker test and are attending an appointment to receive your results, you should prepare a list of questions to ask your physician, using the 'Questions to ask **after** biomarker testing' below as a guide. You should aim to use your appointment with your physician to gain a full understanding of your test results and their implications for your treatment going forwards.

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

Below are examples of questions you should ask your physician before and after receiving a cancer biomarker test [13, 14, 15]:

### Questions to ask **before** biomarker testing

- What is the goal of the cancer biomarker test I am receiving?
- What mutations or abnormal processes is the test looking for?  
What would these mutations indicate about my cancer?
- Which specific cancer biomarkers should be tested for my tumour? Why?
- Will I be receiving single- or multi-biomarker testing?
- **[If single biomarker testing]** Why is multi-biomarker testing not being performed?
- Where will my sample be collected?
- Will I need more than one biopsy?
- Where will the test be carried out?
- How will the test be done?
- How reliable is the test?
- What is the cost of this biomarker test? Is it covered by my insurance?
- **[If testing is not covered]** What are my options for receiving funding for the test? Are you able to support me in receiving funding?
- How long will it take to get the results of this test?
- How do I know if I am a potential candidate for a clinical trial?
- Where can I get information about clinical trials?
- Is there any additional information I should have before receiving the test?





### Questions to ask *after* biomarker testing



- Which biomarker tests were performed, and what were the results?
- Can I get a copy of my test results?
- *[If the results are negative / unclear]* Should I be retested?
- How could my test results affect my course of treatment?
- What treatment approach do you recommend, and why?
- Are there any approved medicines that target my specific cancer subtype based on my test results?
- Do these results tell us anything else about my prognosis?
- Are there any clinical trials open to me based on these results?
- Are there other cancer biomarker tests that should be performed now or in the future?
- Do the results suggest that biomarker testing is recommended for any of my family members?
- Is there any additional information I should have following the outcome of my test?

## 13. How to get additional resources and support

The cancer patient organizations in your country, as well as your health insurance agency can be a good source of information. You may also consult the ECPC members in your country (<https://ecpc.org/about-us/members/>). Other sources of information can be cancer hospital websites and the cancer medical associations in your country. ECPC publishes during the year relevant information about biomarkers

Organisation	Website
 European Cancer Patient Coalition (ECPC)	<a href="https://ecpc.org/biomarkers-and-patients-access-to-personalized-oncology-drugs-in-europe/">https://ecpc.org/biomarkers-and-patients-access-to-personalized-oncology-drugs-in-europe/</a>

## 14. Glossary of common biomarker cancer testing terminology

Below is a glossary of terms you may come across in the context of biomarker testing:

Glossary	
<b>Cancer biomarker</b>	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. Biomarker refers to any molecule in the human body that can be measured to assess health (e.g., mutation in one of the BRCA genes as an indicator of breast or ovarian cancer risk). Molecules can be derived from blood, body fluids or tissue [3,4]
<b>Biomarker-linked therapy</b>	Therapy for which a biomarker test is specified in the medicine label; the presence or absence of the biomarker determines whether a patient is eligible to receive the therapy
<b>Biomarker testing</b>	A measurement developed to detect and/or quantify one, or several, cancer biomarkers for the diagnosis and/or prognosis of cancer patients. Tests can be divided into three groups: chromosome tests (looking for abnormal changes within the structures that contain your DNA), gene tests (assessing a particular part of your DNA for changes; assessing gene expression changes), and biochemical tests (assessing the presence of abnormal proteins or specific chemicals in blood, other body fluids or tissue) [3,4]
<b>Companion diagnostic</b>	Test that is linked to the use of a particular treatment
<b>Comprehensive testing</b>	The evaluation of tumour or blood samples to detect multiple alterations in genes that are known to drive cancer growth. Includes next-generation sequencing (NGS)
<b>Health technology assessment (HTA)</b>	Systematic evaluation of the properties, effects and / or impact of a health technology (e.g., a cancer biomarker test). Multidisciplinary process to evaluate the social, economic, organisational and ethical issues associated with a health intervention or health technology. The main purpose of conducting an assessment is to inform decision-making (e.g., whether a test should be reimbursed) [18]
<b>Liquid biopsy</b>	Sampling and analysis of liquid biological tissue, primarily blood. Also known as fluid biopsy or fluid phase biopsy. Like traditional biopsy, this type of technique is mainly used as a diagnostic and monitoring tool for diseases such as cancer, with the added benefit of being largely non-invasive (blood sample is used for the test as opposed to a patient's tumour biopsy)

<b>Molecular diagnostics (MDx)</b>	Collection of techniques used to analyse cancer biomarkers in order to diagnose and monitor disease, detect risk and aid therapy selection; examples include PCR (polymerase chain reaction) and NGS (next generation sequencing)
<b>Next generation sequencing (NGS)</b>	Large-scale DNA sequencing technology in which millions of nucleotide sequences are deciphered simultaneously. Allows for sequencing the entire genome (whole genome), all known genes (whole exome) or only selected genes (target panel)
<b>Polymerase chain reaction (PCR)</b>	Technique used to amplify small segments of DNA. Once amplified, the DNA produced by PCR can be used in many laboratory procedures, including the diagnosis of cancer
<b>Precision medicine (PM)</b>	Or <i>personalized</i> medicine; a healthcare approach that utilises molecular information and health data from patients to generate insights to prevent or treat disease [5]. Referred to as precision oncology in the context of cancer
<b>Single biomarker testing</b>	Test evaluating the presence of a single gene mutation or expression of a single protein within a biopsy associated with a particular form of cancer (e.g., HER2 testing in breast cancer patients)
<b>Test technologies</b>	Methods used to perform biomarker tests; includes immunohistochemistry (IHC), fluorescence in situ hybridisation (FISH), polymerase chain reaction (PCR), next generation sequencing (NGS)

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