Personalised Medicine: A Guide for Patients

An Initiative of the
European Cancer Patient Coalition

European Cancer
Patient Coalition

CRACKING THE
CANCER CODE
PERSONALISED MEDICINE
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Preface

Prof Mark Lawler, Scientific Committee ECPC

Personalised medicine is rapidly becoming an important diagnostic and therapeutic approach in many human diseases. Cancer researchers and cancer doctors have led the way, so that there are now a significant number of personalised medicine treatments that have reached the clinic, some of which are delivering superior outcomes for cancer patients across Europe. However, personalised medicine has a number of complexities when compared to existing treatment approaches. It is important that the cancer patient is well informed as to the opportunities and challenges that personalised medicine presents, so that they can make the best and most informed decisions about their personal care.

In order to provide information on some of the opportunities that personalised medicine can provide and address some of the challenges that it presents, the European Cancer Patient Coalition decided to create a resource that would specifically address the needs of patients. "*Personalised Medicine: A Guide for Patients*" has been co-created by patients and health professionals and seeks to provide relevant information on personalised medicine for cancer patients and address questions that the cancer patient might have in relation to this relatively new field of medicine. It is intended to inform patient of the opportunities that personalised medicine can bring, while also acting as an empowerment tool for cancer patients in making what can often be difficult decisions about their care.

Understanding the details of personalised medicine and how it might help specifically in your care may be challenging. Additionally, there may be a number of uncertainties that you have, but find it difficult to articulate to your doctor or to ask the right questions. To help you in this regard, we have created a series of 10 questions that you as a patient may choose to ask your doctor, which hopefully will ensure that you are more informed so that you can make the decision that is right for you.

Personalised medicine has the potential to deliver significant benefit for cancer patients. We intend this guide to be a patient-focussed enabler that you can employ to allow you to make the best decision that is right for you and enhances your personal health and wellbeing.

Prof. Lawler is Associate Pro-Vice-Chancellor, Professor of Digital Health and Chair of Translational Genomics at Queen's University Belfast, UK. He is a member of the Scientific Committee of ECPC, Member of the Board of the European Cancer Organisation and Member of the Board of the European Alliance for Personalised Medicine

Glossary

BCR-ABL: a protein that is specific to patients with certain forms of leukaemia (all Chronic Myeloid Leukaemia patients and a subset of patients with Acute Lymphoblastic Leukaemia). Detection of this protein in the blood or marrow of patients indicates that they may be suitable for a specific personalised medicine therapy.

Companion Diagnostics: Diagnostic tests that indicate a particular treatment that a patient should receive – it is important that both the diagnostic test and the subsequent treatment are available to the patient and are that both are reimbursed (paid for) by the health system.

Epidermal Growth Factor Receptor: A particular protein whose activity is changed in a number of different cancers. Knowing whether the activity is changed or not allows selection of a particular personalised medicine treatment for the patient.

***erbB2* gene:** A gene that is overactive in a number of cancers, particularly an aggressive form of breast cancer.

General Data Protection Regulation: A regulation created by the European Union which governs the use of data, including health data.

Herceptin: A drug that specifically targets the overactivity that is associated with the *erbB2* gene (see also *erbB2* gene).

Immunotherapy: A treatment approach for patients with cancer that specifically aims to activate the patient's own immune system against the cancer in their body.

Liquid Biopsies: Blood samples taken from patients that contain biological features of the tumour which can be detected and used as biomarkers to select the optimal therapy for the patient.

PDL-1: One of a number of proteins which can be detected within the patient's biopsy or blood sample and used to decide whether an immunotherapy treatment can be used in that particular patient.

***Ras* oncogene:** A gene that is altered in different cancers (e.g. colorectal, lung cancer) and may be employed as a biomarker to decide which treatment a patient should receive.

Real Word Evidence (RWE): The data that are collected, both during and after treatment and which may be used to help inform the development of future treatments. Real World Evidence studies are becoming increasingly more common as they look at how a drug is performing in the real world (i.e. in the clinic post approval of the drug), rather than looking at it solely in the clinical trial setting.

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Main author
Prof. Mark Lawler

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What You need to know about Personalised Medicine

1. Personalised Medicine: A knowledge-based approach to treat your tumour

1.1 Research delivers new knowledge

In the last 20 years, our understanding of human disease has been amplified by a global research effort to determine the key biological events within our bodies that lead to the development of particular diseases e.g. cancer, heart disease, diabetes.

1.2 Why is this new knowledge relevant to you?

Applying this knowledge has been a key driver for better patient care. In cancer, this "intelligence" has been translated into new diagnostic tools and innovative treatment approaches, initially in clinical trials, but increasingly as standard-of-care, leading to potentially longer, healthier lives for cancer patients. This translation of knowledge to more accurate diagnosis, coupled to more precise treatment that targets your particular tumour, is known as "personalised medicine". The European Cancer Patient Coalition strongly feel that cancer patients must be well informed of their options, as articulated in the First Article of the European Cancer Patient's Bill of Rights.[1] In this booklet, we provide relevant information about personalised medicine, empowering you to make informed decisions about your care.

[1] https://ecpc.org/policy/cancer-patients-rights/

2. Why, What and How – answering your questions about personalised medicine

2.1 Why Personalised Medicine?

In the past, cancer patients received much of their treatment through a "one size fits all" approach, where all patients (with say breast cancer) were given essentially the same treatment. However, the greater understanding that research has provided of the specific biological characteristics of cancer have revealed that malignancies like breast cancer are not a single disease. There are many different subtypes of breast cancer, which differ at the biological level. This facilitates a more focussed diagnostic and treatment approach, specific to your particular tumour. Targeting your tumour in this way may also lead to less treatment side effects.

2.2 So what is personalised medicine?

In the context of cancer, personalised medicine is a treatment approach tailored specifically to certain biological features of your tumour. As this therapy has been designed to target your particular tumour subtype, its chances of success are potentially higher than a more traditional non-targeted approach e.g. chemotherapy. Personalised medicine aims to deliver to you "the right treatment, at the right dose, at the right time," thus maximizing the chances for this personalised treatment to work for you, controlling your disease and preserving your quality of life (Figure 1).

2.3 How Personalised Medicine?

Personalised medicine is more complex than standard cancer care approaches. Ideally, it is best delivered in recognised cancer centres, with all the relevant specialized staff incorporated into a multidisciplinary team (MDT). In the MDT, your particular diagnostic work-up and treatment is discussed and agreed, and this discussion should include your opinions and preferences. Laboratories should have the most optimal diagnostic equipment.

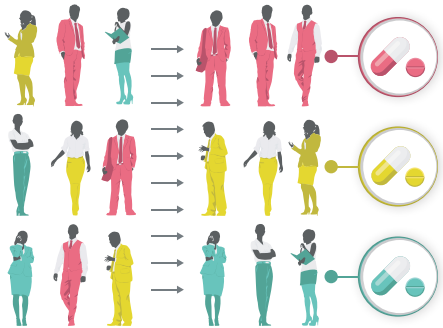

Figure 1

3. Getting personal: Cancer Biomarkers and Targeted Treatment

3.1 Cancer Biomarkers

The first part of your personalised medicine journey involves a specific diagnostic test that will be performed on your blood or tumour biopsy sample. This will determine what personalised medicine treatment is most suitable for you. These tests are called cancer biomarkers or companion diagnostics, detecting specific feature(s) within your sample that characterize your individual tumour. Liquid biopsies, where characteristics of the tumour are picked up within the patient's blood without the need for a tissue sample, are also being used.

3.2 Targeted treatment

Once the cancer biomarker has indicated which treatment option is the best for you, your MDT will deliver your specific treatment. The MDT brings together all relevant expertise required to manage your specific cancer and will include oncologists, radiotherapists, oncology surgeons, cancer nurses, pathologists. molecular biologists, geneticists, technologists and other relevant specialists, depending on your specific disease.

Depending on the results of your biomarker test, there may be more than one option for treatment, so you will have the chance to discuss your options with your MDT. The treatment may involve targeting of a gene or protein that is characteristic of your particular tumour, or of a biological pathway that is in some way altered in your cancer. The key feature of personalised medicine is the specificity of the treatment approach, such that tumour cells are targeted for elimination while normal cells and tissues in your body are spared from any potential treatment side effects. The clinical team aim is both to treat your tumour successfully, but also as far as is possible to preserve your quality of life. All options and scenarios must be discussed with you so that your choices are clear (including your option to be treated in another centre) and your expectations are managed.

4. Personal Cancer Medicine options for different types of cancer

4.1 Chronic Myeloid Leukaemia: The Personalised Cancer Medicine exemplar

If you have a cancer such as Chronic Myeloid Leukaemia (CML), your tumour will contain a protein called BCR-ABL that is specific to your disease. Knowledge of this biological feature of your tumour has facilitated the design of a drug that specifically targets this tumour-specific protein and is highly effective in shutting down its activity (Figure 2). Detecting this protein allows the clinical team to select this specific treatment for you.

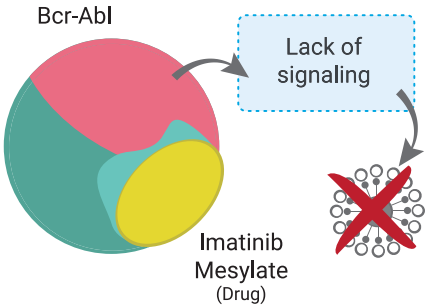

Figure 2

4.2 Personalised medicine for other cancers

If you have lung cancer, testing for the epidermal growth factor receptor (EGFR) which is implicated in lung cancer development, will allow the clinical team to determine whether your tumour can be treated with a therapy that targets this protein. If you have colorectal cancer, determining what *ras* oncogene you have in your tumour may open up a more precise treatment option. If you have breast cancer, you may be one of the approximately 30% of breast cancer patients who have an overactive *erbB2* gene. Testing for this overactivity will allow you to be treated with a drug such as Herceptin, which targets this cancer protein and has been shown to be a highly effective therapeutic approach.

4.3 Immunotherapy; personalised cancer medicine for all?

More recently, stimulating your immune system to fight your cancer (the promising treatment avenue of immunotherapy) offers potential for certain tumour types, so you may be tested to see for example if your PDL-1 gene is active. PDL-1 is one of a number of cancer biomarkers that can determine if an immunotherapeutic intervention is suitable for you (Figure 3). However, while immunotherapy approaches have shown promise, much research still needs to be done before it becomes a standard-of-care.

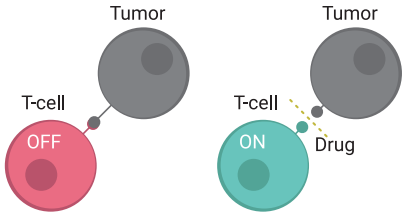

Figure 3

5. Coping with treatment

5.1 Potential side effects of your treatment

Although personalised medicine treatment may in general lead to fewer side effects, certain complications of treatment may occur and persist. It is important that you are made aware of this possibility prior to commencement of your treatment.

5.2 Development of Resistance to Personalised Medicine treatments

While personalised medicine offers a more focused approach to treatment, it is important to realise that cancer is a complex disease. Thus, while you might respond to the initial treatment, your tumour may become resistant to that treatment over time, leading to the reemergence of your cancer. The development of 2nd and 3rd generation personalised medicines for particular tumour subtypes has enhanced our ability to combat treatment resistance and these newer drugs may be employed in your current or future treatment. However, it is important to realise that resistance to treatment cannot be addressed in all circumstances or for all cancers.

6. Personalised medicine research – can you contribute?

6.1 Research as an enabler for better patient care

Research is a vital component of our ongoing fight against cancer. Evidence indicates that patients treated in research-active centres have better outcomes than those who are not. A critical component of the research effort in personalised medicine is the enrollment of patients in cancer clinical trials or real world evidence (RWE) studies. These studies facilitate development of more accurate cancer biomarkers and more effective treatments. However, it is important that your consent as a patient is sought (and received) for the use of either your samples or your data if they are being collected as part of any research study. Increasingly, patients are also being empowered to have a more central role in the research itself, with opportunities to contribute their knowledge to the evolving research effort. The best cancer research considers not only the views and expertise of scientists and healthcare professions, but also the experience and knowledge of patients.

6.2 The importance of data

The data generated from research studies that you may have decided to participate in, will provide crucial evidence to help underpin better outcomes for cancer patients. Your data can make a great contribution to this evidence, but the collection and effective use of your data must be performed in a responsible fashion. You must have consented to the use of your data for research and your privacy must be protected and respected, respecting the General Data Protection Regulation (GDPR). The question of anonymization is an important one; while it guarantees anonymity, this must be balanced against your potential wish to have information about the characteristics of your tumour returned to you, which may be relevant for future diagnostic tests.

6.3 Patient participation in cancer research

You should be informed about the potential to participate in research studies, as research is the way by which better diagnostics and more effective treatments for patients are delivered. However, it is important to realise that while the conduct of personalised medicine research may lead to more accurate diagnosis or more effective cancer treatment, these results may not impact on your particular treatment, but be of benefit to future cancer patients. Thus, your participation in this research, either through analysis of biopsy or blood samples that you donate, or evaluation or sharing of your data that you consent to be used, can be an altruistic activity, but one that the scientific and medical community deeply appreciates. However, it is important for you to be informed that your tissue/blood sample or data may be used in subsequent research studies and that you are asked to give permission for this future use.[3] It is important for you to be reassured that it is your choice as to whether you participate in a research study or not, that your wishes in relation to the research that you give your permission to be involved in are respected, and most importantly, that if you don't wish to participate in a research study, this decision will not in any way affect the quality of your care.

[3] Details on sample donation, biobanking and consent in cancer research can be found at https://ecpc.org/wp-content/uploads/2019/08/ecpc_eurocanplatform_biobanking_faqs-2.pdf

6.4 Barriers to personalised medicine

Although personalised medicine has a role to play in the care of cancer patients, there are still barriers to be overcome. Cost can be an issue – many of the newer personalised medicine therapies are expensive, so it is important to check that your potential treatment costs are covered by your health system or will be reimbursed by your insurer. Also, your cancer treatment and its companion diagnostic must both be made available. Often the paradoxical situation arises that the treatment may be reimbursed but the laboratory test that is employed to decide the most suitable treatment for your tumour is not. If this is the case in your county or region, you should look to lobby with your local or national patient advocacy group so that you get access to both the test and the treatment.

7. Preparing for your medical appointment

7.1 The Informed Patient

In order for you to negotiate the challenges and complexity that personalised medicine can pose, it is important that you become an informed patient, with clear information at hand that allows you to understand how a personalised medicine approach will be applied to your particular cancer. Your right to information is captured within the European Code of Cancer Practice[2] a patient-centred manifesto which details what you should expect from your health system. Information on personalised medicine treatment options should be provided to you in clear language that you can understand and be supported by additional information from reliable sources that gives you a fuller appreciation of what a personalised medicine approach will mean specifically for you. Providing this information and support should help you in making your decision to receive (or not to receive) a personalised medicine tailored for your particular tumour type.

[2] https://www.europeancancer.org/2-standard/66-european-code-of-cancer-practice

7.2 Assisting you to make an informed decision

As part of being an informed patient, it is important for you to have the opportunity to ask specific questions about your personalised medicine journey. However, in many situations, you may struggle to appreciate what are the key questions to ask. In this section, we provide a series of **10 questions** that you should consider asking of your doctor or multidisciplinary cancer team, in order to allow you to make a fully informed decision on receiving (or not receiving) a personalised medicine treatment approach .

7.3 Ten Questions that you should ask your doctor or medical team

1. Are there personalised medicine treatment options for my particular tumour?

This is a key starting question, so that you know your options at the start of your personalised medicine journey

2. Is this treatment option available for me at this hospital?
It is important to know if the treatment that is best for you is available at your local hospital, and if not, are there options for you to be treated elsewhere in your region/country

3. I want to get access to a personalised medicine treatment. What is involved?
This is another key question so that you are well prepared to make your personal decision.

4. If my test indicates that a specific personalised medicine treatment is suitable for me, what happens next?
This allows you receive more information to help make your decision on whether this treatment is both suitable for you and acceptable to you.

5. If the test indicates that the specific treatment is not suitable for me, do I have other options?
This scenario can be challenging; sometimes there will be other options but in other situations these options may not exist. It is important that your expectations are managed, so that you have a clear appreciation of your options.

6. Is my treatment part of a clinical trial or is it a standard-of-care?
It may be that the only option for a personalised medicine for you is through a clinical trial. A full description of what is involved should be provided and you should be consented to participate in this clinical trial and for your data to be used (if that is your wish).

7. How often has this hospital treated patients using this personalised medicine approach and what do their results look like compared to other hospitals in my region/country?
It is important to be reassured that your hospital has relevant experience in the delivery of the particular personalised medicine treatment to patients and that their levels of success are similar to other hospitals in your region/country.

8. Are there any side effects associated with the personalised medicine that I will receive and if so what are they and how will they be managed?
Increasingly as a patient, you balance your decision on receiving a particular treatment based on whether it will successfully treat your tumour, against your desire to preserve (in as much as is possible) your quality of life. So it is important for you to know what side effects (if any) you should expect, and how these will be managed.

9. How (and when) will I know that my treatment has worked?
Particular tests may be required to see whether your treatment has worked or not. It is important to know how long it will take for results to be relayed back to your doctor (and you)

10. Can you signpost me to reliable information/ resources that will help me make my decision and keep me informed of the latest personalised medicine developments?
There is a large volume of information about personalised medicine but it may be difficult for you to appreciate its accuracy and reliability. Having access to reliable information and to patient advocacy organisations that will support you will be important, both in your decision making but also in reassuring you on your personalised patient journey. ECPC will signpost patients to the most relevant, reliable and up-to-date information and provide support through national organisations affiliated to ECPC.

We hope that these 10 Key questions will support you as a patient in navigating the often complex and confusing world of personalised medicine. Our guide is an evolving one and we encourage you to work with us and let us know of your experiences so we can ensure that our guide remains current, informative and supportive to all cancer patients across Europe.

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