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| Pan-Nordic clinical consent framework for genetic testing Included within this document you will find the following to aid in the delivery of informed consent in combination with a healthcare provider to patients in the clinical context for genetic testing; for use with a non-vulnerable, adult population:   1. Guidance document to the below documents 2. Genetic testing information sheet 3. Genetic testing consent form   These documents were developed as part of a joint project between the Nordic Alliance for Clinical Genomics (NACG) and Nordic PerMedLaw to develop Nordic harmonized clinical consent documents for genetic testing. More information is available on the NACG website, here:  <https://nordicclinicalgenomics.org/projects/nacg-pan-nordic-consent-project> |

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| Date | 04.01.2020 |
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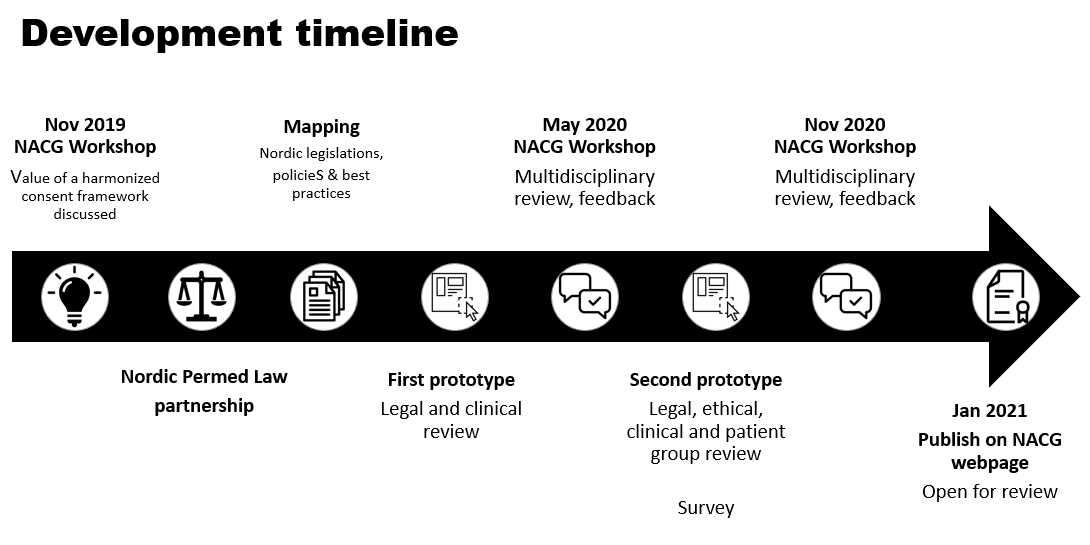
# Guidance to the genetic testing information sheet & consent form

Please refer to the following guidance regarding the genetic testing information sheet and consent form prior to adoption in your healthcare institution. The documents may require multiple adjustments to tailor it to your healthcare institution before use with patients. The genetic testing information sheet and consent form do not represent professional or legal advice of any kind. Obtaining informed consent from patients participating in genetic and genomic testing in the clinical context requires legal and ethical review prior to administration to patients. As a result, healthcare institutions should seek appropriate legal guidance when developing their consent processes.

The purpose of the genetic testing information sheet and consent form are:

1. to aid in the delivery of informed consent in combination with a healthcare provider to patients in the clinical context for genetic testing; and
2. to be used with a non-vulnerable, adult population.

Background development:



The genetic testing information sheet and consent form were developed by the Nordic Alliance for Clinical Genomics (NACG), the BIGMED legal network and, following their establishment in May 2020, the Nordic PerMedLaw legal network. The work was led by DNV GL, Precision Medicine research program who are NACG members.

As of today (22 Dec 200), in the Nordic countries, there is no single, standardized approach to administering informed consent to patients undergoing genetic testing. Starting January 2020, NACG conducted a Nordic multi-disciplinary focused approach to developing a harmonized approach to clinical consent framework and toolkit development with the following motivation:

1. As the leading precision medicine initiative in the Nordics, NACG is well-positioned to initiate and co-ordinate discussions around consent practices across the Nordic countries in genetic testing.
2. Development of a harmonized consent framework can serve as a vehicle to harmonize and identify categories for discussion in consent in genetic testing and data sharing across the Nordic countries.
3. Development of a harmonized consent framework can enable partnerships across disciplines and borders in consent in clinical genetic testing

In brief, the development process included multiple stages that started with a comprehensive mapping of Danish, Islandic, Finnish, Norwegian, Swedish and European legislation, best practices, and policy on the topic of informed consent and genetic testing in the clinical context. A multi-disciplinary network consisting of those working in healthcare institutions in genetic testing (such as genetic counselors, laboratory personnel, clinicians and healthcare leaders), legal advisors specifically from the Nordic PerMedLaw network, patient groups and industry partners working in quality assurance and genetic testing all provided input to on-going reiterations to the consent documents over the course of a year. The input was gathered through the following channels:

1. Two major workshops in May 2020 and Nov 2020, consisting of approx. 50-60 participants in total.
2. Three review sessions followed by reiterations by NACG and Nordic PerMedLaw legal network members for input via google document for edits and comments.
3. Interviews and surveys to stakeholder groups such as clinicians, patient advocacy groups, laboratory and healthcare leaders in genetic testing.

# General guidance

1. Review each section to ensure that the content is complementary to your healthcare institution.
2. The documents are in *Word* and therefore provide opportunity for desired edits at any area of the form to be customized to your institution.
3. Specific sections in the genetic testing information sheet are customizable. For example, among these sections you are given several statements to choose from and these can be edited. Ensure that the options you do not choose are subsequently deleted. Ensure that the headings highlighted in blue are also deleted.
4. The documents are designed to be modifiable to include the inclusion of the healthcare institution’s logos and name. The NACG logo may be removed.
5. The documents are recommended to be translated into local languages. An authorized/certified translator is advised.

# Guidance to the genetic testing information sheet

The genetic testing information sheet is meant to be used within the setting of a healthcare institution in combination with a healthcare provider to aid in the process of delivery of informed consent to patients undergoing genetic testing. The information sheet can be used as a guide to walk patients through the various aspect of genetic testing.

1. **What is genetic testing**

This section is meant to inform the patient to what genetic testing is and examples to how it is performed.

1. The purpose of genetic testing

This section is meant to inform the patient to the purpose of genetic testing.

1. What you will be tested for

This section is meant to inform the patient to what they will be tested for. This section requires adaptation to the healthcare institution by selecting an option provided or editing the text. Make sure to delete the text that does not apply.

1. Benefits and risks to genetic testing

This section is meant to inform the patient of the benefits and risks of genetic testing. This section is meant to encourage the patient to raise any questions or concerns with their healthcare provider. Sections provide information to the A. benefits and B. risks and limitations and can be populated with more text if needed. The risks and limitations section includes the following topics: No findings or variant of uncertain significance (VUS) and unexpected family relationships.

1. Implications of genetic testing

This section is meant to inform the patient that even though a genetic test may confirm a diagnosis, there may be no intervention or treatment available. This section informs the patient that further testing may be required to confirm the diagnosis.

1. Secondary and/or incidental findings

This section is meant to inform the patient of secondary and incidental findings and their right to choose whether these results are returned to them or not. Secondary findings are described as clinically actionable or non-clinically actionable, and drop-down options are provided for the healthcare institution to indicate if the patient will have the option to be notified or not of these findings. This section requires close attention to ensure it aligns with your healthcare institution’s management policy of secondary and incidental findings and provides a “free text” section where details can be included that may not be offered in this section. This section aligns with the consent form and thus requires careful attention to ensure that these sections are consistent. Ensure that the other options which do not apply are deleted.

\*This section does not include information on ACMG59, which may be included in the free text option here.

1. Voluntary nature of the test

This section is meant to inform the patient of the voluntary nature of the test. This section includes information on right to know and right not to know. This section includes an option for the healthcare institution to inform the patient on what will happen to their data if a patient decides not to be told their genetic testing results. Ensure that the other options which do not apply are deleted. Note that the consent form aligns with this section where consent is requested. At this section, a “free text” section where details can be included that may not be offered in this section are provided.

1. Information relevant for relatives

This section is meant to inform the patient that a finding of a hereditary or a predisposition to a disease may have implications for their relatives. This section includes an option for the healthcare institution to inform the patient to the process of notification of relatives and their rights. Options are provided to choose from. Ensure that the other options which do not apply are deleted. Note that the consent form aligns with this section where consent is requested. At this section, a “free text” section where details can be included that may not be offered in this section are provided.

1. Delivery of results

This section is meant to inform the patient of the approximate timeline for the laboratory to receive their sample, processing, analysis and interpretation. Indicate in the blue section the amount of time. This section is also meant to inform the patient of the approximate timeline for when they can expect to receive their results. Indicate in the blue section the amount of time.

1. Reanalysis

This section is meant to inform the patient of potential future reanalysis possibilities for their data. A section is provided to modify this section with options to choose from that will inform the patient of the healthcare institute’s procedures for reanalysis. Ensure that the options which do not apply are deleted. The last paragraph informs the patient to how long their data is stored and requires this information to be provided by the healthcare institution.

1. Data sharing

This section is meant to inform the patient to the purpose of sharing their personal data, how and where it is shared, and how personal data is defined. This section requires careful attention to ensure that two sections are adapted to the healthcare institution. The first section provides options to choose which healthcare data may be shared. Ensure that the options that do not apply and are not selected are deleted. The second section is a free text option where the healthcare institution can specify where personal data will be shared, and information about with? which countries or geographies, which healthcare institutions, and which databases. Note that the consent form aligns with this section where consent is requested.

1. Research

This section is meant to inform the patient that they can choose to be contacted to submit their anonymized data for research. Note that the consent form aligns with this section where consent is requested. An option is provided to modify this section with options to choose from that must be edited and that will inform the patient of the healthcare institutions’ procedures for research inclusion. Ensure that the options that do not apply and are not selected are deleted.

1. Contact for questions

This section is meant to provide the patient with contact options if they have questions outside of the visit with their healthcare provider / genetic counseling. It is recommended that email and telephone options are provided.

# Guidance to the genetic testing consent form

This form is meant to follow discussions the patient has had with their healthcare provider allowing them to voluntarily express their consent for genetic testing.

1. Introduction

This section is meant for the patient to confirm that they have read the information sheet, spoken to a healthcare provider and received information about the genetic test protocol, what their results can contain, and how their data will be handled. The patient will also confirm if they have had enough time to consider the consent form, had the opportunity to discuss genetic testing and its implications, been given access to information about genetic testing, and been able to ask questions until satisfied.

1. About the test

This section is meant for the patient to indicate they understand the primary purpose of their genetic test, benefits, risks, and their right to choose not to receive the results. The patient also indicates they understand test results may change and procedures are in place to review knowledge against patient data to identify opportunities for updated diagnoses. This section includes options in blue text to choose if several genes, the whole exome or whole genome will be sequenced. Ensure those that do not apply are subsequently deleted. This section is according to the healthcare hospital policy (indicated in sections 1-7 of the information sheet). If the patient has understood the information in this section, they are asked to consent to the genetic testing by providing their initials.

1. Potential outcomes

This section is meant for the patient to indicate that they have been informed of the potential outcomes of the genetic test. This section aligns with sections 3, 4, 5, & 6 in the information sheet. If the patient has understood the information in this section, they are asked to consent to the genetic testing by initialing this section.

1. Information relevant for relatives

This section is meant to obtain consent for sharing of their genetic results data in the situation that a finding of a hereditary or predisposition to a disease that may have implications for their relatives according to the healthcare hospital policy (indicated in section 8 of the information sheet). The patient is asked to provide consent by initialing this section.

1. Secondary findings

This section is meant to obtain consent for the return of secondary and / or incidental findings to the patient according to the healthcare hospital policy (indicated in section 6 of the information sheet). The patient is asked to provide consent by initialing this section. This section includes options in blue text to choose between clinically actionable and/or non-clinically actionable in text and initialing box. Ensure those that do not apply are subsequently deleted

1. Permission to share your data for diagnostic purposes

This section is meant to obtain consent for the sharing of their personal data according to the healthcare hospital policy (indicated in section 11 of the information sheet). This section also includes an option to provide additional text (e.g. type of data, where it is shared). The patient is asked to provide consent by initialing this section. The standardization of this section across the Nordic countries may enable data sharing and thus benefit patients.

1. Permission to be contacted for research

This section is meant to obtain consent to be contacted about future research opportunities. This section is according to the hospital policy (indicated in section 12 of the information sheet). The patient is asked to provide consent by initialing this section. This section includes options in blue text to choose between being contacted about or participating in research in the initialing box. Ensure those that do not apply are subsequently deleted.

1. Consent

This section is meant to obtain consent for the primary purpose of genetic testing. The patient is asked to provide consent by providing their name and information required by the hospital.

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| Genetic testing information sheet Your healthcare provider has recommended a genetic test for you. The following information is meant to support the discussion you will have with your healthcare provider to ensure that you have understood the purpose and significance of a genetic test. |

## About the test

### What is genetic testing

A genetic test can help identify if there is a change in your genetic material that may cause a disease. The test is usually performed on a blood sample, cheek swab or tissue.

2. THE PURPOSE OF GENETIC TESTING  
The purpose of a genetic test is to try and identify the cause or risk of a suspected disease by analyzing your genetic material for an abnormal change (variant).

### **3. What you will be tested for**

Select for your healthcare institution:

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| --- |
| **A.** A single gene / variant responsible for a specific, suspected genetic disease, or multiple genes (gene panels, whole exome or whole genome sequencing) in parallel. |
| **B.** Several genes, your whole exome or whole genome will be sequenced in order to determine a gene variant or variants responsible for a specific, suspected genetic disease. |
| **C.** Free text |

### 4. Benefits and risks to genetic testing

Taking a genetic test is your choice. Therefore, it is important that you have discussed and understood all the information that you have been given to help you make your own decision. It is also important that you can discuss with your healthcare provider any questions or worries that you may have. Genetic testing can bring great benefits, but there are also possible risks and limitations. Some of the benefits and risks are discussed below. The list is not complete, and not all the points will be relevant to your specific situation. They might however give you some useful things to think about and discuss with your healthcare provider.

### A. Benefits

A correct diagnosis may predict the course of your disease and affect the choice of treatments and screening programs relevant to you.

### B. Risks and limitations

It is possible that knowledge from the test results may result in psychological stress for you and your family. It is always recommended you discuss the results with your healthcare provider.

### No findings or variant of uncertain significance (VUS)

Knowledge about genetic disease is still incomplete. It is possible to receive results without findings, which means that with our current knowledge, we cannot find the genetic cause, while uncertain findings mean we do not understand their significance yet. Future testing and/or knowledge may help clarify this.

### Unexpected family relationships

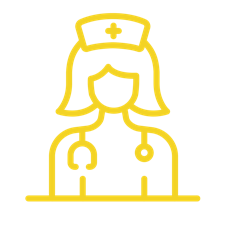
The results from genetic testing may identify unexpected family relations, for example adoption or paternity.

### 5. Implications of genetic diagnosis

Even though a genetic test may confirm a diagnosis, there may be no intervention or treatment available. Depending on the results and laboratory procedures, further testing may be required to confirm the diagnosis.

### 6. Secondary and/or incidental Findings

Genetic analyses, particularly those involving a large number of genes, may identify secondary and/or incidental findings that are not directly related to the initial reason for your testing. However, such findings could still be of medical importance for you and your family, as they may provide information about a risk that you may not be aware of for potentially serious, unavoidable genetic diseases.

As an example, clinically actionable findings are those in which a genetic change is identified that reveals or predicts a genetic condition for which preventative measures and/or treatments are currently available. These could be related to your initial purpose for seeking a genetic test or could be a secondary finding. For example, a recessive hereditary (i.e. not preventable) disease (such as cystic fibrosis) or a hereditary genetic mutation identified as a hallmark for increased cancer risk (such as BRCA1/2 in breast and ovarian cancer).

Free text (delete this when complete):

|  |
| --- |
| Free text |

If a secondary finding is clinically actionable, you will:

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| A. Be notified. |

If a secondary finding is NOT clinically actionable, you will:

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| B. not be notified. |

Secondary and/or incidental findings will not be shared without your consent, unless ethically required or allowed by law.

### 7. Voluntary nature of the test

The genetic test is voluntary. In the case that you decide not to take the test, you should consult with your healthcare provider for possible alternatives, and implications for diagnosis and treatment.

Your right-to-know and not-to-know is protected by law, and our procedures are in line with this. This means that you have the right to know, or not to be informed about test results (right not to know).

Select (one or more) for your healthcare institution:

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| **A.** You can change your mind about having genetic testing or being told the results. |
| **A.** You can choose not to be told the result after the test is finished, but the test result will be placed in your medical record. |
| **C.** Free text |

### 8. Information relevant for relatives

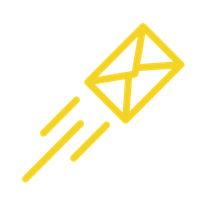
A finding of a hereditary or a predisposition to a disease may have implications for you and your relatives.

In this situation,

Select (one or more) for your healthcare institute:

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| --- |
| **A.** Your relatives may be referred for genetic testing. |
| **B.** Your specific genetic analysis may be shared across genetic services to ensure that the correct testing can be offered to your relatives. |
| **C.** You may be asked to share your genetic testing results to your family members to ensure that the correct testing can be offered to your relatives. |
| **D.** This can be done in coordination with you, or alternative efforts will be made to ensure that your identity is not revealed to those family members. |
| **E.** Free text |

### 9. Delivery of results

Once the laboratory has received your sample, processing, analysis and interpretation can take between [X-X] days.

Results will be received by your healthcare provider and the implications of these will be shared with you in approximately [X] weeks’ time.

### 10. Reanalysis

If your genetic test does not identify a cause for your disease your results may be subject to reanalysis later.

Reanalysis is:

Select (one or more) for your healthcare institute:

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| **A.** Triggered by new genetic knowledge that may impact your genetic test results. |
| **B.** Triggered by routine reanalysis (approximately every 12 months) |
| **C.** At the request of your healthcare provider. |
| **D.** At your request to your healthcare provider. |

Your sample will be stored at the testing laboratory for [X] years, to enable further testing or support quality and result verification, in accordance with standard procedures.

### 11. Data sharing

Personal data consists of information that can be linked specifically to you. Your personal data will be handled according to the EU General Data Protection Regulation (GDPR) and national laws, and sharing of this data is carried out with security safeguards in place.

Your personal data may include:

Select (one or more) for your healthcare institute:

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| **A.** Your raw unprocessed data |
| **B.** Your genetic sequence with variants annotated (interpreted results of the genetic analysis) |
| **C.** Your associated health information related to the purpose of the testing |
| **D.** Your original biological material and resulting processed sample |

The sharing of your genetic data and related health information may aid in obtaining a diagnosis for yourself. The processing of your personal data may include sharing via other databases and laboratories to identify what your test results mean by comparison to other people. In a reciprocal manner, the sharing of your results may help another patient receive a diagnosis. However, this sharing may provide no direct benefit to yourself or your family. Your personal data will not be shared without your consent, unless required or allowed by law.

If you consent, your personal data may be compared to other patients’ results across:

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| Free text: (for example, which countries or geographies, which healthcare institutions, which databases?) |

### 12. Research

Select (one or more) for your healthcare institute:

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| **A.** For long term storage and research purposes your sample/data will be automatically enrolled in (e.g. biobank name) |
| **B.** Your options to modify your consent for this must be carried out through (e.g. contact details) |
| **C.** You have the opportunity to consent to be contacted about participation in research on the consent form. |

### 13. Contact for questions

Provide (one or more) for your healthcare institute:

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| Free text area... |

|  |
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| Consent for genetic testing This form is meant to follow discussions you have had with your healthcare provider allowing you to voluntarily express your consent for genetic testing. |

# Consent for genetic testing

*This form is meant to follow discussions you have had with your healthcare provider allowing you to voluntarily express your consent for genetic testing.*

## It is my choice to have genetic testing.

1. I confirm that I have read the information sheet, spoken to a healthcare provider and received information about the genetic test protocol, what my results can contain, and how my data will be handled. I have had enough time to consider this consent form and have:

* had the opportunity to discuss genetic testing and its implications.
* been given access to information about genetic testing.
* been able to ask questions until I am satisfied.

### About the genetic test

I understand that:

* the genetic test will attempt to identify the cause of a suspected disease by analyzing my genetic material for an abnormal change (variant). Several genes / the whole exome / the whole genome will be sequenced in order to determine a gene variant or variants responsible for a specific, suspected genetic disease.
* if I change my mind, I can choose not to be told about the results.
* tests are based on current best-practice knowledge, and this knowledge may change in the future.
* the implications of my results may change at a later time point.
* procedures are in place to review knowledge against patient data to identify opportunities for updated diagnoses.

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| **Initial here** if you understand the information presented above about the genetic test and consent to the testing |  |

### Potential outcomes

I understand that:

* there are no guarantees that this test will find a cause for my condition(s).
* the significance of my results may be uncertain or unknown; meaning an identifiable variant is not (yet) relatable to a disease diagnosis.
* unexpected family relationships may be identified.
* further testing or information sharing may be needed to verify the results.

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| **Initial here** if you understand these are potential outcomes to genetic testing and consent to the testing |  |

### Information relevant for relatives

I understand that:

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| --- | --- |
| **Initial here** if you consent to have your results shared with relatives if a hereditary or a predisposition to a) disease are identified. |  |

* a finding of a hereditary or predisposition to a disease may have implications for my relatives.

### Secondary findings

I understand that:

* the results may identify clinically actionable and/or non-clinically actionable secondary findings, not related to the initial reason for my genetic test.

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| **Initial here** if you consent to being informed about clinically actionable and / or non-clinically actionable secondary findings if they are identified. |  |

### Permission to share your personal data for diagnostic purposes

I understand that:

* sharing of my genetic data and related health information may aid in obtaining a diagnosis for myself and for others. However, this sharing may provide no direct benefit to myself or my family.
* a preference not to share my personal data will not affect the service I receive.
* my personal data that will be processed includes:

*Option to fill in for your healthcare institute:*

|  |
| --- |
| Free text area (e.g., type of data, where it is shared) |

|  |  |
| --- | --- |
| **Initial here** if you consent to sharing your personal data as indicated above. |  |

### Permission to be contacted about RESEARCH

The opportunity to participate in research that may or may not be related to my reason for undergoing genetic testing may arise. These research opportunities may or may not provide any direct benefit to my health or treatment. All research will be approved by regulatory and ethical boards prior to being conducted.

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| **Initial here** if you consent to be contacted about / participate in future research opportunities. This will entail the sharing of your personal contact information (e.g. name and email). |  |

### Consent

I understand that my genetic material will be tested for changes in genes that currently are known to be associated with my condition. I consent to having genetic testing as summarized in this form.

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| --- | --- | --- | --- |
| **Patient signature** |  | **Healthcare provider signature** |  |
| **Date** | DD.MM.YYYY | **Date** | DD.MM.YYYY |
| **Patient identifying number** |  | **Healthcare provider** |  |