

# Genetic testing information sheet

*Your healthcare provider has recommended a genetic test for you. In order to ensure that you have understood the purpose and significance of a genetic test, we are providing you with the following information about the test.*

## About the test

### WHAT IS GENETIC TESTING

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

### THE PURPOSE OF GENETIC TESTING

The purpose of a genetic test is to identify the cause of a suspected disease by analyzing your genetic material (DNA) for an abnormal change (variant).

### WHAT YOU WILL BE TESTED FOR

A single gene/variant responsible for a specific, suspected genetic disease, or multiple genes (gene panels, whole exome or whole genome sequencing) in parallel.

### BENEFITS AND RISK 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.

### BENEFITS

A correct diagnosis can predict the course of your disease, affect the choice of treatments and screening programs, as well as making it possible to understand the implications of this disease for your family members.

### RISKS AND LIMITATIONS

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

### NO FINDINGS OR VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)

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

### 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.

### SECONDARY FINDINGS

Genetic analyses, particularly those involving a large number of genes, may identify secondary findings that are not directly related to the actual reason for your testing. However, such findings could still be of medical

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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 or non-treatable genetic diseases.

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 available. These could be related to your initial purpose for seeking a genetic test or could be an unrelated finding. For example, a recessive hereditary 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).

Select your healthcare institute:

Text

If a secondary finding is clinically actionable, you will:

Choose an item.

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

Choose an item.

### VOLUNTARY NATURE OF THE TEST

The genetic test is voluntary. In the case that you refuse to take the test, you should consult with your healthcare provider for possible alternatives, and implications for diagnosis and treatment.

Select (one or more) for your healthcare institute:

- A. You can change your mind about having genetic testing or being told the results.
- B. You can cancel the test at any time before the laboratory finishes the test.
- C. You can also choose not to be told the result after the test is finished, but the test result will be placed in your medical record.

D. You have the right to erasure, that is having your data erased.

### INFORMATION RELEVANT FOR RELATIVES

Your genetic results will not be released to your family members. However, your relatives may be referred for genetic testing, if a clinically actionable hereditary diagnosis is identified. In this situation, your specific genetic analysis may be shared across genetic services to ensure that the correct testing can be offered to your relatives. This can be done in coordination with you, or alternatively efforts will be made to ensure that your identity is not revealed to those family members.

### POLICY AND PRACTICES AROUND RIGHT TO KNOW OR NOT TO KNOW

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) and to stop the testing processes that have been started at any time up to being given the results and to request the destruction of all analysis results.

Populate for your healthcare institute:

Free text area...

### 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 health provider and the implications of these will be shared with you in approximately [X] weeks' time.

### REANALYSIS AND RECONTACT PERIOD

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

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Reanalysis is: *Select (one or more) for your healthcare institute:*

**A.** Triggered by new genetic knowledge that may impact your genetic test results.

**B.** Triggered by routine reanalysis (approximately every **[how often]**)

**C.** At the request of your healthcare provider.

**D.** At your request to your healthcare provider.

**A.** Your raw unprocessed data

**B.** Your genetic sequence with variants annotated (interpreted results of the genetic analysis)

**C.** Your associated health information

**D.** Your original biological material and resulting processed sample will not be shared but may be used for further /future testing to confirm your diagnosis or to understand your results.

## DATA SHARING

A preference not to share your data will not affect the service you receive, however there may be an increased likelihood of obtaining a diagnosis if you do so.

## PURPOSE OF SHARING

Your healthcare provider may share both your unprocessed results, genetic variants and health information with 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. Sharing genetic data and associated health information can advance our knowledge about genetic diseases.

To decide whether findings are significant for yourself or others, your personal data may be compared to other patients' results across: **Free text:** *(prompt: for example, which countries or geographies, which healthcare institutions)*

## TYPE OF DATA

Sharing of your data is carried out with security safeguards in place. These include the removal of personal identifiers (such as your name, address, and personal ID number) and using secure restricted-access databases. Personal data that may be shared includes:

*Select (one or more) for your healthcare institute:*

## CONTACT FOR QUESTIONS

*Option to fill in:*

Free text area...

## GDPR-STATEMENT

Your personal data will be handled according to the EU General Data Protection Regulation (GDPR). **[NAME OF HOSPITAL HERE]** is responsible for the handling of your personal data and only healthcare staff will have access to your data.

In accordance with GDPR you have the right, which you can exercise at any time, to:

- > be provided with information about what data exists and where it is stored
- > make rectifications to incorrect data
- > have your data erased
- > withdraw or modify your consent with effect for the future

This can be done by contacting **[INSERT NAME HERE]**.