HIRSLANDEN PRECISE

INFORMATION FOR PATIENTS

# INFORMED CONSENT TO GENETIC TESTING

**The aim of genetic testing is to detect or exclude a genetic disease or a predisposition to it by analysing the genome. This sheet contains important information on what you should consider before undergoing a genetic test. You will also receive information on the areas of application, the background and the methodology of genetic testing.**

## Clarification before a genetic test

Genetic testing is voluntary and requires your formal consent, as it provides very personal results. In order for you to be clear about the possibilities, consequences and limits of genetic testing, we recommend that you seek genetic counselling from relevant specialists before undergoing such a test.

We also recommend that you take sufficient time to discuss all the questions that concern you, so you can make an uninfluenced personal decision. Below are some key points that should be discussed with you before you decide for or against genetic testing:

- Importance of genetic testing for medical care, including diagnosis, progression, prevention and treatment options for the disease being investigated.
- Possible significance of genetic test results for other family members.
- Significance of the test; indication of the possibility of an inconclusive or unexpected outcome.
- Risk of an unfavourable outcome, as well as decisions and consequences that may result for you (including any impact on your insurance coverage).
- Alternatives to genetic testing.
- Your right to refuse the proposed test. Decision on what to do with your test material after the test has been conducted: storage for possible reuse, archiving, use for medical research or destruction.
- Collaboration with our partner laboratory, Unilabs, for the analyses.
- Information on the processing of your personal, patient and medical data.
- Information on the costs, confirmation of cost coverage and whether your health insurance company will cover the costs or not.

## Procedure

A small amount of venous blood or oral mucosa cells from a cheek swab are usually used for genetic testing. Blood samples need not be taken on an empty stomach. If a cheek swab is planned, you should not drink or eat anything for 1 hour beforehand so sufficient oral mucosa cells can be obtained. Occasionally, other tissues are also used for genetic testing (e. g. skin, hair, muscle).

## Legal basis

In Switzerland, the Federal Act on Human Genetic Testing (HGTA, SR 810.12, BBl 2018 3509) and its implementing ordinances provide the legal basis for genetic testing.

## Areas of application of genetic testing

Today, genetic testing provides important information in almost all areas of medicine. The applications can be divided into the following main areas:

**Diagnostic** tests are used to genetically diagnose or confirm an existing medical issue. They often allow a definitive diagnosis with corresponding significance for treatment.

**Pre-symptomatic** and **predictive** tests make it possible to determine whether a healthy person carries the predisposition to develop a certain disease.

**Prenatal** tests are used to detect or exclude genetic diseases in the foetus. A distinction is made between non-invasive tests (e. g. first trimester screening [FTS], non-invasive prenatal testing [NIPT]) and invasive prenatal testing, such as amniocentesis or chorionic villus sampling (CVS).

**Examinations are carried out on family members** to determine whether a person carries a genetic change that is already known in the family and can be passed on to their offspring. Often, the person examined does not show any symptoms of the disease.

**Somatic tests** determine whether genetic changes are present, e. g. in cancer tissue. The primary aim is to obtain information on the therapy and prognosis of the cancer; however, a family predisposition to cancer may also come to light, for example.

## Background and methodology of genetic testing

The human genome is located in the cell nucleus of every single body cell (from red blood cells) and consists of DNA. The genome is organised into 23 pairs of chromosomes, one pair of which contains the sex chromosomes (XX in women, XY in men). Around 20 000 genes are lined up on the chromosomes and form the basic units of the genome. A few genes are located in the cell outside the cell nucleus in the mitochondria. Together, all the genes form the blueprint for the structures and metabolism of the body. Changes in this blueprint can cause illness. There are changes that will very likely trigger a disease, as well as changes that only increase the risk of disease.

As a rule, genetic changes are sought at two different levels:

1. **Chromosome level:** Changes in the number or structure of chromosomes (chromosomal abnormalities). Large chromosomal abnormalities are usually detected by microscopic chromosome analysis. Small chromosomal abnormalities can also cause severe disease patterns; however, they can only be detected with high-resolution chromosome analysis (array CGH).
2. **Gene level:** Disease-causing changes in the DNA sequence of genes (gene mutations). Gene mutations are detected by various molecular methods. The search for a gene mutation can focus on a specific gene, or it can analyse numerous genes or even the entire genome at the same time (high-throughput sequencing). If the entire genome is examined, it is very difficult to interpret the data. When many genes are analysed simultaneously, there is also an increased risk of identifying changes in genes that are not directly related to the clinical issue or the disease being sought (incidental findings) or that may or may not be clinically significant, i.e. disease-causing, or not.