

# PRECISE CANCER

REDEFINING HEALTH



# PRECISE CANCER

Every year around 41 700 people in Switzerland develop cancer (predominantly breast, prostate, lung, bowel or skin cancer). However, there are now more than 200 different known forms of cancer. The causes of cancer are extremely complex – for example, the environment and cell metabolism can be responsible for the disease. However, in five to ten per cent of cases, an increased risk of cancer is caused by hereditary gene variants (germline mutations). Moreover, age or extreme exposure to harmful substances are also common reasons for cancer.

Due to the enormous developments in genetics and genomic medicine as well as the advances and new findings in the early detection and treatment of cancer, a new research and speciality area has emerged: Oncogenetics. The aim of oncogenetic examinations is:

- to identify the relevant germline mutation in patients who are suspected of having hereditary cancer and to initiate appropriate treatment.

- to determine the risk that healthy relatives from families with proven hereditary cancer will develop the disease. Appropriate preventive measures can then be recommended depending on the findings.

## PERFORMANCE AREA

## WHICH GENES ARE BEING EXAMINED?

There are more than 300 different known tumour-associated genes today. These are mainly genes that are involved in the process of repairing genetic information (DNA), cell division, cell growth and cell death.

### Base genes

The following genes have been scientifically well researched and are examined on the basis of the clinical question:

#### Breast and ovarian cancer:

*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*

#### Prostate cancer:

*ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, TP53*

#### Lung cancer:

*ATM, BRCA1, BRCA2, CHEK2, EGFR, TP53*

#### Stomach cancer:

*APC, CDH1, CHEK2, KRAS, MLH1, MSH2, MSH6, MUTYH, PRKARIA, PMS2, TP53*

#### Bowel cancer:

*APC, BMPR1A, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*

#### Pancreatic cancer:

*APC, ATM, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PALLD, PMS2, STK11, TP53, VHL*

#### Skin cancer:

*BAP1, BRCA2, CDK4, CDKN2A, EPCAM, MC1R, MITF, MLH1, MSH2, MSH6, PMS2, POT1, PTEN, RB1, TP53*

For information on genes that are associated with the investigation of a therapy or drug intolerance, please refer to the document "Precise Pharma".

## Special genes

The progress in oncogenetics is astounding. New gene variants are constantly being discovered and their meaning explained. This can make it necessary to determine other genes in addition to the basic genes. The selection of the corresponding genes is made either in the first oncological or oncogenetic consultation or in a second step after the initial results have been discussed with the oncological or oncogenetic specialist.

Depending on the clinical constellation, a gene panel may also be proposed. A large number of genes are examined, including those that are not exclusively or not directly related to the underlying oncological disease. Accordingly, it is possible that the experts come across other gene variants by chance. Therefore, patients are informed about

this possibility in advance and can decide in advance whether they would like to be informed about such incidental findings. Alternatively, they can exercise their right not to know and waive this option with a signed authorisation.

If there is a gene panel, the focus is on the cancer genes relevant to the current clinical question. It is possible to analyse other genes with the same DNA sample at a later date, and receive timely results.

## TARGET GROUP

### WHO IS ONCOGENETICS AIMED AT?

- Patients with cancer at a young age
- Patients with more than one type of cancer
- Patients with bilateral cancer in paired organs (e.g. breast, ovaries)
- Patients with cancer that has occurred in other family members (for example, breast cancer, bowel cancer, prostate cancer)
- Patients with cancer from families in which other family members have a combination of certain types of cancer (e.g. breast cancer and ovarian cancer, bowel cancer and uterine cancer)
- Healthy people from families with frequent cancers, especially those at a younger age

**PROCEDURE**

# HOW DOES A GENETIC ANALYSIS WORK?



## Registration

Done by the family doctor or a specialist.

STEP  
**01**



STEP  
**02**

## Preliminary discussion & approval

Before the test, a preliminary genetic discussion takes place to determine the indication and the genes to be examined. Written consent from the patient is mandatory.

### Genomic board

If necessary, the cases are discussed by specialists from a variety of disciplines and recommendations are made.



## Sampling

A blood sample is taken by qualified personnel and sent to the laboratory. Depending on the situation, buccal swabs can also be used.

STEP  
**03**



STEP  
**04**

## Laboratory analysis

The samples are analysed in the certified laboratory with specialised genetic methods and visualised on the computer with the help of bioinformatic programmes. The analysis time is between 1 and 4 weeks, depending on the genetic diagnosis.



## Evaluation & reporting

The commissioning doctor receives the results in the form of a written report. The patients receive a copy of the report after the findings have been discussed.

STEP  
**05**



STEP  
**06**

## Genetic advice & individual treatment recommendations

The expert in genetic medicine explains the analysis results in an interview so that there are no misinterpretations. On the basis of the genetic results, further care is provided by the family doctor or specialist. Depending on the situation, further individual diagnostic and/or therapeutic measures are suggested.

## BENEFITS

# WHY IS GENETIC TESTING WORTHWHILE?

Evidence of pathogenic gene variants:

- confirms the diagnosis,
- allows a more precise prognosis of the course of the disease,
- enables reliable clarification of further family members (especially first-degree relatives),
- allows a specific choice and early start of therapy and
- improves the acceptance of a (possibly lifelong) therapy.

If the genetic finding is normal, this makes a diagnosis rather unlikely, but does not rule out such a diagnosis with absolute certainty. The detection of therapy-relevant gene variants allows the use of patient-specific drugs with the best possible effect and as few side effects as possible.



Comprehensive support from a specialist



Simple sampling by taking blood or a buccal swab



Faster test results



High accuracy and security

## PRICE & CONDITIONS

The costs of a genetic examination depend to a large extent on the complexity of the analysis and range from a few hundred to several thousand Swiss francs.

Genetic analyses are covered by compulsory health insurance if they are classified as mandatory. However, to do this, they must be used to diagnose or treat an illness and its consequences (Article 25 paragraph 1 KVG). Specifically, this means that there is an acceptable probability that they will result in at least one of the following consequences:

- Decision on the necessity and type of medical treatment
- Directional change in the medical treatment used so far
- Directional change in the necessary examinations (e.g. for the timely prevention, detection or treatment of typically expected complications)

- No further examinations of typically expected symptoms, secondary diseases or complaints

Analyses in which it is already clear at the time of the order that the result does not have any of the consequences mentioned are excluded from the assumption of costs and must be borne by the people who want a genetic test. The genetic counselling, like any other medical service, is billed via Tarmed. The tariff for the position "genetic counselling" corresponds to the basic tariff for a general medical consultation.

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