

PRECISE CARDIO

REDEFINING HEALTH



PRECISE CARDIO

Cardiovascular diseases are the leading cause of death in the industrialised world. To date, research and clinical practice have concentrated heavily on the identification and management of risk factors. However, in future, the focus will be primarily on the disease-specific cause and its explanation on the basis of the genetic background and environmental factors.

Research into human genetic material has developed significantly over the past few years. In this way, gene variants that are important for clinical heart medicine could be identified. This new branch of research is called cardiogenetics. The following gene groups can be analysed using cardiogenetics:

- Genes associated with a patient's existing heart disease.
- Genes associated with a family member's heart disease.
- Genes that are associated with drug therapy and increase the risk of intolerance or heart damage.

PERFORMANCE AREA

WHICH GENES ARE BEING EXAMINED?

There are more than 300 different known genes associated with heart diseases today, the majority of which are genes that regulate the contraction capability of the heart muscle or that control the heartbeat.

Base genes

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

Cardiac arrhythmias (Brugada syndrome, Long QT syndrome and others):

ANK2, CACNA1C, CACNB2, HCN4, KCNE1, KCNE2, KCNH2, KCNJ5, KCNQ1, SCN1B, SCN3B, SCN4B, SCN5A

Heart muscle diseases (cardiomyopathies):

ACTC1, BAG3, CRYAB, DES, FLNC, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTR

Coronary heart disease:

ABCA1, APOB, APOE, LDLR, LDLRAP1, PCSK9

Coagulation disorders:

F2, F5, PAI1

For information on genes that are associated with the diagnosis of a therapy or drug intolerance and/or increase the risk of damage to the heart, please refer to the document "Precise Pharma".

Special genes

The progress in cardiology is astounding. New gene variants are constantly being discovered and their meaning clarified. As a result, it may be necessary to determine other genes in addition to the basic genes and depending on the clinical question. The selection of the corresponding genes is determined either in the first cardiogenetics consultation, or in a second step after the initial results have been discussed with the cardiological 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 directly related to the underlying cardiological disease. Accordingly, it is possible that the experts come across other gene variants by chance. Therefore, patients are informed about this pos-

sibility 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 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 CARDIOGENETICS AIMED AT?

- Patients with genetic heart disease
- Patients with suspected genetic heart disease
- People with cardiac disease in the family
- People with suspected inheritable heart disease in the family
- Family members of patients with sudden cardiac death <45 years of age
- Patients with cardiac arrhythmia
- Patients with coagulation disorders and a tendency to thrombosis
- Patients with drugs that are pharmacogenetically relevant, especially drugs that are toxic to the heart (e.g. cancer therapies)

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.

Hirslanden Precise

hirslanden.precise@hirslanden.ch
+41 44 386 45 44