swissdnalysis
step up your quality of care

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SWISS ACCREDITATION
SMTS 0060

Patient Name / First Name:
Date of Birth:
Sex: **Phone:**
Street: **ZIP/City:**
Insurance: **Insurance No.:**

GENERAL INFORMATION ON GENETIC TESTING FOR USERS

SwissDNAlysis stands for molecular genetic diagnostics at the highest level. It is important to us that you understand the principle of our genetic testing as well as what to expect in the genetic test results. Relevant information about this test can be found on these pages. Please take your time to read it carefully. We are happy to answer any further questions.

Genetic Counseling

A genetic consultation is recommended before and after performing genetic testing. As part of this genetic counselling, the individual seeking advice should have answered all the questions by the physician recommending the test. You should receive the following information: the precise description of the disease to be tested, why it is important to perform the genetic test, implications of the test for you, how the test will change medical decisions, prognosis or therapy, implications of the test for your family, how the particular disease in question is inherited and eventual risks for relatives. Detailed information will be provided to you about molecular genetic testing of the disease in question, its benefits and significance, and the interpretation of results.

Frequently Asked Questions:

What are genetic diagnostics and why is it performed for hereditary diseases?

A genetic test is a medical test in which a person's genetic material is analysed for DNA changes, usually to diagnose a hereditary disease or to assess the risk of a specific condition. This supports the attending physician in diagnosis and therapeutic decision-making. In certain cases, molecular genetic diagnostics also allows improved counselling regarding disease progression and prognosis. Targeted testing for specific risks can also be offered to family members who can benefit to be identified before symptoms occur.

What is required to perform the genetic test?

1. We can extract the genetic material (DNA) from many types of tissue. Ideally, we use 3–5 ml of blood. Sometimes other types of tissue can be used, including kidney, liver, heart buccal swab, among others.
2. Genetic testing is voluntary. A fully completed and signed consent from the patient or legal representative is a legal requirement (see page 3). During a detailed consultation, you will be informed about the purpose of genetic diagnostics, what happens during analysis, and the potential implications for you and your relatives.
3. Approval from health insurance or confirmation of cost coverage (cost approval) is required.

What happens to my sample and genetic data after testing?

The data obtained will be processed in accordance with the Swiss Federal Act on Data Protection (DSG) and the Federal Act on Human Genetic Testing (GUMG, GUMV). Data security and confidentiality are our top priorities. Unless otherwise requested, blood or tissue samples are stored for 1 year, extracted DNA is stored for 5 years, and the reports and associated data are archived for at least 10 and up to 20 years.

To support scientific progress, anonymized results may be included in public databases or published in peer-reviewed journals.
The results of the test are communicated to the referring physician and to any physicians designated by the patient. Without consent of the person tested, **NO** test results will be shared with anybody.

How long does the genetic test take?

Turnaround time depends on the test: NGS or panel tests will take around 15 days to 8 weeks (average 45 days).
Single variant or family testing (cascade screening): 3–15 days (average 5 days).

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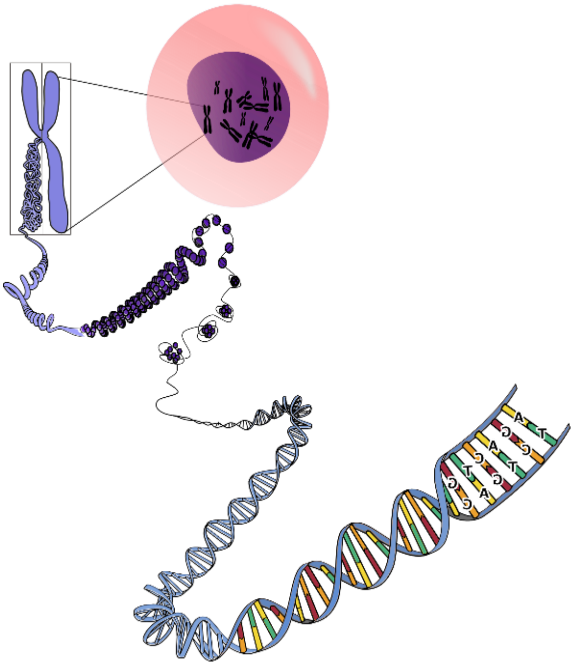
What is a mutation / variant?



The genetic material contains the blueprint or instruction to build our body, encoded or stored in genes. A gene is a segment of DNA that provides instructions for protein synthesis. Humans have approximately 20,000–25,000 genes. The genetic field understand the function of less than 10,000 genes. Genes are part of another structure called chromosomes which are stored within the cell nuclei. A change in the DNA sequence (the genetic code made by “letters” represent the four chemical building blocks of DNA A=Adenine, G=Guanine, C=Cytosine, and T=Thymine) is called a genetic variant or mutation. Some genetic variants can change how a protein is made or how well it works. Because proteins are important for many body functions, these changes may sometimes cause a disease or increase the chance of developing certain health conditions. However, most genetic variants do not affect health. Many are simply part of normal human differences, and some variants may even help protect against certain diseases.

Each person has two copies of most genes—one inherited from their mother and one from their father. Genetic conditions can occur depending on whether one or both gene copies are affected. **Autosomal recessive conditions**: A person usually develops the condition only if both copies of the gene have a disease-causing variant. Individuals with only one affected copy are typically called “carriers” and usually do not have symptoms. **Autosomal dominant conditions**: A disease-causing variant in just one copy of the gene is enough to cause the condition. Genetic testing may sometimes identify unexpected findings that are unrelated to the original reason for testing. These “incidental findings” may provide information about other health conditions or future health risks. You can choose whether or not you would like to receive this type of information. Incidental findings are relatively uncommon, especially in targeted Gene-panel tests that we perform at SwissDNAlysis.

What is the principle of diagnostic NGS testing?

Our genetic panel testing uses Next-Generation Sequencing (NGS), an advanced technology that allows many genes to be analysed at the same time. Variants considered clinically important are confirmed using an additional method called Sanger sequencing to ensure accuracy. Genetic variants are interpreted and classified according to current professional and scientific guidelines. As scientific knowledge continues to evolve, the interpretation of some variants may change over time. In addition, genetic testing cannot detect all disease-causing changes, and new disease-associated genes or genetic regions may be discovered in the future. Some results may be uncertain or may require further testing or clinical evaluation for clarification. The interpretation of genetic findings is based on the best available scientific knowledge at the time of testing.

Because genetic information is shared within families, some findings may also have implications for relatives, who could have an increased risk of developing the same condition or of carrying the same genetic variant.

We report three main types of genetic variants:

Pathogenic variants: There is strong scientific evidence that the variant causes disease.

Likely pathogenic variants: Current evidence strongly suggests that the variant is disease-causing, although some uncertainty may remain.

Variants of Uncertain Significance (VUS): There is currently not enough scientific information to determine whether the variant is harmless or disease-causing. These variants may not have been previously reported or studied in enough detail. In such cases, re-evaluation after 2–5 years may be recommended, as new scientific information may become available over time. In some situations, testing other family members —especially relatives who are affected by the condition—can help clarify whether a VUS is related to the disease. This is called genotype–phenotype correlation or family segregation analysis.

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INFORMED CONSENT AND REQUEST TO PERFORM GENETIC TEST

This form must be completed by the referring physician and the person to be tested.
After answering ALL questions, send this page to SwissDNAlysis.
Missing information or signatures will delay the test results.

I confirm that I have received genetic counselling, and I understand the need to perform a genetic test. I had sufficient time to consider my decision. All my questions have been answered.

1. I give my consent to perform the following analysis (*Please choose Panel OR Single mutation test*):

☐ **Gen-Panel (NGS)** (To test one or more genes; panel descriptions are available online: www.swissdnalysis.ch)

For the following disease: ____________________

Report *CYP2C19*-*2,*3-Status? ☐ No ☐ Yes
(*Optional for Clopidogrel or Mavacamten treatment cases*).

☐ **Single Mutation Test:**

Gene 1: ____________ **DNA-Position:** c.____________ **Protein-Position:** p.____________

Gene 2: ____________ **DNA-Position:** c.____________ **Protein-Position:** p.____________

In which relative was the original variant identified? ____________________

2. The following doctor should receive a copy of the genetic test report:

____

3. In case of no insurance coverage:

☐ Do not perform the test.
☐ Perform the test (patient bears cost).

Is this test funded by research / institutional funds?

☐ No ☐ Yes: Which? ____________________

4. Does the patient have symptoms?

☐ No ☐ Yes: Please attach medical report or describe the symptoms:

____
____
____

5. Sample type submitted to be tested:

☐ EDTA-Blood ☐ Buccal swab ☐ Extracted DNA ☐ Other tissue: ____________

6. Date of sampling: ________/________/________

7. I give my consent for:

To use my sample for quality control (anonymized):
☐ No ☐ Yes

To use my sample for research purposes (anonymized):
☐ No ☐ Yes

To use my clinical data for research purposes (anonymized):
☐ No ☐ Yes

To be informed about incidental findings:
☐ No ☐ Yes

Signature** Patient or legal responsable person* ***Date:	***Signature and Stamp** Doctor* ***Date:***

Free cancellation is possible within one week after receipt of this consent, please contact SwissDNAlysis AG directly.

Laboratory use only
Received on: ______/______/______ **Entered by:** **Checked by:** **Lab No.:**