# GENETIC TESTING INFORMATION IN ACCORDANCE WITH THE GERMAN GENETIC DIAGNOSTICS ACT (GenDG)

Dear Patient,
A genetic test has been recommended for you or a person under your care to address certain health concerns. This test is being carried out in accordance with the requirements of the **Genetic Diagnostics Act (GenDG).** The law mandates that you are informed about the procedure before any genetic analysis is conducted and that you provide **written consent.**

Under the GenDG, the physician who orders a genetic test for medical purposes is considered the responsible party. According to the GenDG, any physician, regardless of specialized qualifications, is authorized to order a diagnostic genetic test.

## PROCEDURE OF THE GENETIC TESTING

**Purpose of the test:**
The genetic analysis aims to identify alterations in your genetic material that may be responsible for your symptoms or conditions, or for diseases that are already known in your family.

**Methods of testing:**
Various methods can be used for genetic testing, such as chromosome analysis (cytogenetic analysis) or DNA analysis (molecular genetic analysis). The best-suited method will be chosen by the laboratory based on the specific question to be answered. In some cases, a combination of methods may be used to increase the likelihood of detecting a genetic alteration.

**Conducting the test:**
The test is typically carried out using a blood sample. The venipuncture generally poses no health risks.

## POSSIBLE RESULTS OF THE GENETIC TEST

1. **Identification of a disease-related genetic alteration:**
   - If a (likely) pathogenic genetic alteration is identified that is linked to your condition, this indicates that the alteration is most likely the cause of your symptoms or condition.
   - You will be provided with detailed information about the condition, including its progression and treatment options, if available.

2. **No identification of a disease-related genetic alteration:**
   - Even if no disease-related genetic alteration is found, a genetic cause for your condition cannot be ruled out.
   - This is due to the fact that not all clinical abnormalities can be genetically explained, as environmental factors may also play a role. Additionally, it is possible that the genetic cause could not be detected by the methods currently used.
   - For this reason, a follow-up consultation and possibly a re-evaluation in 2-4 years is recommended if the results are unremarkable.

3. **Identification of genetic alterations with unclear clinical significance:**
   - Sometimes, genetic alterations are identified whose clinical significance is not yet clear. These so-called variants of uncertain significance (VUS) may be either harmless or disease-related, but their effect is not yet fully understood at the time of testing.
   - For this reason, a follow-up consultation and re-evaluation of the genetic analysis in 2-4 years is recommended for any identified VUS.
   - You will be informed of such genetic alterations, and discussions will be held with you regarding their potential implications for your health and the health of your family.

4. **Incidental findings:**
   - **Definition:** In comprehensive genetic tests, such as exome sequencing, incidental findings may occur. These involve genetic alterations unrelated to the original reason for testing
   - **Example:** In a test for a developmental disorder, a genetic predisposition to a heart arrhythmia may also be discovered.
   - **Your decision:** You may specify in the consent form whether you wish to be informed of incidental findings. Typically, only those incidental findings are reported that are related to conditions for which medical treatment options are available or preventive measures can be taken.

## CONFIDENTIALITY AND DATA PROTECTION

Your genetic test results will be stored exclusively in a **secure laboratory information system** and are only accessible to **authorized personnel.** All data protection regulations will be adhered to according to the applicable legal requirements. The results must be stored for **10 years**, after which the data will be destroyed unless you have made other arrangements.

## REVOCATION OF CONSENT

You can **revoke** your consent for the genetic test at any time, either fully or partially, without providing any reasons. You can also stop the test and request the destruction of the tested material before the results are communicated. However, any costs incurred prior to your objection may be charged.

## FURTHER QUESTIONS?

If you have any questions about the methods, results, or any other aspects of the genetic test, please feel free to contact your responsible physician.