Information prior to genetic testing according to the Genetic Diagnostics Act (GenDG)

The German Society for Human Genetics (GfH) and the Professional Association of German Human Geneticists (BVDH) point out that the Gene Diagnostics Act (GenDG) requires detailed information and written consent from the patient for all genetic analyses. Additionally, prior to prenatal and predictive analyses, Genetic counseling is required. Please read this patient information carefully prior to genetic analysis and contact us if you have any questions.

You (or a person for whom you have custody or who you look after) have been recommended to carry out a genetic analysis to clarify the following diagnosis/question:

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We would like to explain to you the aim of these analyses, what happens during genetic analyses and what significance the results can have for you and your family.

A genetic analysis aims to examine

• the carriers of the genetic material (chromosomes) using chromosome analysis/molecular cytogenetic analysis,

• the genetic material itself (DNA) by means of molecular genetic or array analysis or

• the products of the hereditary substance (gene product analysis)

in regards to genetic properties that may be the cause of the disease/disorder that you or your relatives have experienced or suspected.

In most cases, a blood sample is used as the **test material**. Normally, taking a blood sample does not pose any health risks. Blood can sometimes accumulate in the area of the puncture site (hematoma) or, extremely rarely, nerve damage can occur. If tissue removal is necessary in your case (skin biopsy, amniotic fluid puncture, chorionic villus biopsy, etc.), you will be informed separately about the risks of sample collection for you and, if applicable, for the child you are expecting. Another risk that can never be completely ruled out is the possibility of a sample mix-up. All measures are taken to avoid this and other errors.

A genetic analysis can be done by

• analysing specific individual genetic characteristics, e.g. in the case of a specific suspicion, (using molecular cytogenetic, molecular genetic or gene product analysis)

• analysing many genetic properties simultaneously in the sense of an overview method (e.g. using chromosome analysis, DNA array, genome sequencing).

Meaning of the results

If a disease-causing characteristic (e.g. a mutation) is detected, this finding usually has a high degree of certainty. If no disease-causing mutation is found, mutations responsible for the disease may still be present in the gene examined or in other genes. A genetic disease or predisposition to a disease can therefore usually not be ruled out with complete certainty. In this case, we will try to estimate the probability of the occurrence of the above-mentioned disease or a predisposition in you or your relatives. Sometimes gene variants are detected whose meaning is unclear. This will then be stated in the report and discussed with you. A comprehensive explanation of all conceivable genetic (partial) causes of the disease is not possible. It is also not possible to rule out every risk of disease for yourself or your relatives (especially for your children) through genetic analyses.

Almost all analytic techniques can produce results that are not directly related to the actual question, but can still be of medical importance for you or your relatives (so-called additional findings). Especially using overview methods such as array analyses and genome sequencing, additional findings can occur and may indicate increased risks (of which you may not yet be aware) for potentially serious, unavoidable or untreatable diseases. As part of the consent, you can decide whether and under what circumstances you would like to be informed about such additional findings. If several family members are examined, a correct interpretation of the findings depends on the stated family relationships being correct. If the findings of a genetic analysis lead to doubts about the specified family relationships, we will only inform you if it is necessary in order to fulfill our mandate.

Right of withdrawal

You can withdraw your consent to the analysis in whole or in part at any time without giving reasons. You have the right not to know test results (according to your right not to know), as well as to stop initiated test procedures at any time until the result is communicated, including the immediate destruction of all test materials and all results collected up to that point.