

Information concerning genetic testing

Within Germany, genetic testing is regulated by law (Gendiagnostikgesetz) requiring that all implications of the planned genetic testing procedure be explained in full to the patient.

1. **Your physician has recommended** that you (or someone for whom you have custody / power of attorney) undergo genetic testing in order to exclude or confirm the following condition:
2. **Genetic testing** involves analysis of genetic material (chromosomes, DNA, RNA, protein) obtained from body cells (blood cells, skin cells, buccal smear, amniocytes, chorionic villus etc) in order to detect disease-causing genetic alterations.
3. **Routine procedures** may include venipuncture, skin biopsy, amniocentesis or chorionic villus biopsy. These procedures carry only minimal risks (bleeding, infection). Loss of pregnancy following amniocentesis is rare (less than 0.2%). It is somewhat higher following chorionic villus biopsy (1-3%). Mislabeling of specimens or inadvertent human error in handling and processing the specimen is extremely rare, but not zero.
4. **The scope of the genetic analysis** is determined by the clinical situation. Depending on prior clinical information, there can be a focussed search for alterations (mutations) in a given gene or chromosome, or there can be a more comprehensive screening involving parts of or even the entire genome (e.g. karyotyping, array-CGH, whole genome sequencing).
5. **Significance of results.** As a rule, whenever a genetic alteration (mutation, chromosome aberration, altered or missing RNA or protein) is found, a genetic diagnosis can be established. However, a negative result (i.e. failure to detect a genetic alteration) does not completely rule out a genetic basis for the disease in question. Genes other than those tested or mechanisms other than investigated may cause the disease. In some cases, genetic alterations will be found which are considered “variants” rather than disease-causing changes. Their interpretation is sometimes difficult, such that only statistical or empirical information can be derived. It is essential to realize that genetic testing is of limited power. It cannot yield information on each and every disease for which you, your family and your children may be at risk.
6. **Chance findings.** It is important to realize that there is the possibility of unanticipated findings which may or may not be of significance for you or your family. Such chance findings may even include alterations which impose considerable disease risks for you or your family. Unless you request otherwise, you will be informed of such findings only if they entail medical consequences.
7. **Family members and family structure.** The interpretation of genetic test results depends on the completeness and correctness of the family history (pedigree) information. Please be aware that genetic testing will occasionally reveal discrepancies (e.g. non-paternity) which will only be communicated if they are of relevance for the correct interpretation of test results.
8. **Right to withdraw your consent.** You may withdraw your consent anytime without giving reasons. In addition, you are entitled to refuse a genetic test or to refuse being informed of the result of a genetic test. You are also entitled to ask for termination of ongoing testing procedures, including disposal of testing materials, and of any testing result obtained to date.

Signed at (city) date

first and last name

signature