# Information Sheet in Accordance with the Gendiagnostikgesetz (GenDG) [German Genetic Diagnostics Act] (copy for the patient to keep)

UNIVERSITÄTS KLINIKUM FREIBURG
ZKJ ZENTRUM FÜR KINDER- UND JUGENDMEDIZIN

Dear Patient, Dear Parents and Legal Guardians,

You or your child have been recommended to undergo genetic testing (analysis) to evaluate the following diagnosis / health issue:

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**The Gendiagnostikgesetz (GenDG) [German Genetic Diagnostics Act] requires that patients be informed in detail and give written consent before having genetic testing performed. Predictive (regarding future health events) and prenatal (before birth) analyses additionally require prior counseling by a specialist in human genetics. Please read this Patient Information Sheet carefully. It is designed to inform you about genetic analyses (testing). Do not hesitate to ask us any questions you may have.**

This Information Sheet explains to you the purpose of these analyses, what will happen during genetic testing, and what the results may mean for you and your family/relatives.

**The purpose of genetic testing** is to analyze the chromosomes that carry the hereditary material, the hereditary material (DNA) itself or products of the hereditary material (gene product analysis) using specific techniques to identify genetic traits that may be the root cause of your or your relatives' suspected diagnosis.

**The material tested** is usually a blood sample (5 mL, or often less for children). Sometimes, however, we may need to collect some bone marrow or other tissue (e.g., skin, oral mucosa, hair roots).

**Genetic testing** either selectively analyzes individual genetic traits (e.g., if a specific condition is suspected) or at the same time screens a large number of genetic traits (comprehensive screening methods such as whole genome sequencing). The selected method depends on the health issue.

**Meaning of the test results**
If a disease-causing change (e.g., a mutation) is identified, this finding is usually very reliable. If no disease-causing change is identified, the analyzed gene or other genes may still harbor changes that are responsible for the disease in question. This means that a genetic disease cannot be ruled out completely. It sometimes happens that gene variants are found whose significance is unclear. Your doctor will discuss the test results with you. It is impossible to provide comprehensive information about all conceivable causes of disease that may be due (in part) to genetic changes. Nor can genetic analyses completely rule out that you or your relatives (particularly your children) may be at risk of developing disease.

When several members of a family are tested, a correct interpretation of findings will depend on whether the reported biological family relationships are correct. Should genetic analysis findings cast doubt on reported biological family relationships, we will only tell you if this is crucial to achieving the objective of the requested analysis.

Genetic testing (particularly when using screening methods) may produce results that are not directly related to the health issue under evaluation, but might still be medically significant to you or your family/relatives (chance findings, also known as **incidental findings**). You will be told about such abnormalities if these have immediate medical consequences. In the Consent Form below, you can choose whether you want to be told about all incidental findings.

Test results will only be shared with your consent with the persons designated by you.

**Your right to withdraw consent**
You can withdraw your consent to undergo testing in full or in part at any time without giving a reason. You have the right not to be told about test results (right to not know), to stop initiated analytical procedures at any time before being told the result, and to request that all test materials and all results obtained until such time be destroyed.

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# Consent Form in Accordance with the Gendiagnostikgesetz (GenDG) [German Genetic Diagnostics Act]

| Name and address of the institution / stamp | Patient data:<br>Last name First name<br>Date of birth<br>Street address<br>ZIP (post) code Town or city |
|---|---|

**Your/your child's diagnostic workup requires genetic testing that, in accordance with the Gendiagnostikgesetz (GenDG) [German Genetic Diagnostics Act], requires that you be informed in detail and give written consent prior to testing; prenatal (before birth) and predictive (regarding future health events) analyses additionally require prior genetic counseling. Please read this Consent Form carefully and check (tick) your choices below:**

I have received, read, and understood the Information Sheet describing genetic analyses in accordance with the GenDG, and hereby agree that blood and/or tissue collected from me/my child will be tested for genetic changes. Depending on the health issue under evaluation, comprehensive screening methods such as whole genome sequencing may be used. The sole purpose of this testing is to evaluate:

(diagnosis, gene, or test) .....

I agree that the findings of the analysis/analyses may be shared with:

Ms. / Mr.: ……

If applicable, the findings will also be shared with the study director identified when I was told about the suspected diagnosis.

| | |
|---|---|
| I want to be told about results of the genetic analysis only to the extent that this is of practical relevance to myself and my family regarding the above-mentioned health issue. I do not wish to be told about incidental findings.<br>If No, then I wish to be told about any and all incidental findings that may be relevant to myself or my family/relatives. | ☐ Yes ☐ No<br>☐ Yes ☐ No |
| I agree that results of the genetic tests and analyses will not be destroyed (deleted) after 10 years in accordance with statutory provisions, but will rather be kept for up to 30 years so that they will be available to myself or my family/relatives if necessary. | ☐ Yes ☐ No |
| I agree that excess test material will not be destroyed, but may rather be used in encrypted (pseudonymized) form for retesting/verification of the test results obtained, for quality control, for additional testing as part of the diagnostic workup, and for the purpose of improving diagnostic methods.<br>To be able to also use biological materials and associated data for research purposes, excess test material will be added to a biobank. A separate Information Sheet will inform you about this and request your consent. | ☐ Yes ☐ No |

I have been informed about the purpose, nature, scope, and reliability of the requested genetic testing. I have also been informed about the meaning of the test results and the potential consequences these may have for myself and/or other members of my family.
I have been informed that I may withdraw my consent in full or in part at any time without giving a reason and without any penalty. I have also been informed about my rights not to be told about test results (right to not know), to stop initiated analytical procedures, and to request that the test material and all results obtained until such time be destroyed. I have received a copy of the Consent Form.

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Place, date Patient's/Legal representative's signature Doctor's signature