Institut für Experimentelle Hämatologie und Transfusionsmedizin
Venusberg-Campus 1, 53127 Bonn

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IHT
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Patient Information

Dear Patient,

You or a family member may be affected by a suspected inherited disorder of the coagulation system. Therefore, you are receiving this information sheet and will also be informed during a physician consultation about **inherited coagulation or platelet disorders** and the available diagnostic options. The result of the examination may help in making the right decisions for further treatment.

For a large proportion of coagulation or platelet disorders, a **genetic change (DNA mutation)** is considered to be the underlying cause. Such a change can be detected in a **blood sample**. For this purpose, a blood draw of **up to 5–10 ml** is required. Normally, this blood collection poses no health risks. For premature babies, infants, or toddlers, specific risks can be discussed with your pediatrician. In some cases, a smaller volume may suffice.

Your sample will be analyzed using a **next-generation sequencing (NGS)** technique, allowing for simultaneous analysis of multiple genes. If a genetic cause of a coagulation or platelet disorder is identified, you will be invited for a **follow-up medical consultation**.

However, if no pathogenic variants are found, this **does not rule out** the presence of a disease-causing mutation. Pathogenic variants might be present in the tested gene but are not detectable with the chosen method, or may exist in **other genes** not yet known to be relevant for the disease. Therefore, a **negative result does not exclude a genetic cause** (false-negative result). If relevant, you will be advised further.

When multiple family members are being tested, accurate interpretation of results depends on the correctness of the reported familial relationships. If the results raise doubts about these relationships (e.g., paternity), this will only be disclosed if essential for fulfilling the purpose of the examination.

If you do not consent to the storage of the sample for use in **future analyses** (e.g., for family testing or research), any remaining material will be destroyed either immediately upon request or after the legally required retention period.

Our current understanding of the genetic basis of inherited coagulation disorders is still limited, though steadily expanding. In rare cases, our current analysis may yield no conclusive result. With your consent, we would like to conduct **scientific studies** on your genetic material to gain new insights. In the future, new discoveries may justify analyzing your DNA for currently unknown genes involved in coagulation.

If such findings are clinically relevant for you or your family, we will inform you- **if you wish to be informed**. You **also have a right not to know**, i.e., you can decline to be informed of results.

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If new findings emerge in our department (Institute of Experimental Hematology and Transfusion Medicine), we may publish them in scientific journals, but always using fully **anonymized data**, ensuring that your identity cannot be revealed.

You **may withdraw your consent** to the use or storage of your sample for research at any time, without giving reasons and without personal disadvantages. Upon withdrawal, the sample will be destroyed either immediately (if explicitly requested) or after the statutory retention period.

Only tests related to inherited coagulation or platelet disorders will be performed using your or your family members' blood. **No statements about other diseases will be made.**

Your personal data will be stored in a computer-based system for medical diagnostic purposes. For scientific use, the data will be **pseudonymized,** meaning that researchers cannot identify you. Naturally, medical confidentiality and data protection laws apply without limitation.

You may request the results of your tests at any time. You may also attend a clinical counseling appointment, and if needed, genetic counseling and psychosocial support can be provided.

Every item in your consent form may be **withdrawn** at any time, without reason and without disadvantage.

Place, Date

Name, First name (in block letters)

Signature of the patient / legal guardian

This information sheet serves as documentation for the responsible physician. It does not need to be submitted to the laboratory.