Please send to:
Prof. Dr. med. J. Oldenburg
Institut für Experimentelle Hämatologie und Transfusionsmedizin, G43
Abteilung Molekulare Hämostaseologie
Venusberg-Campus 1, 53127 Bonn

Lab Tel.: (+49)- 0228-287-19429
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Results: PD Dr. B. Pezeshkpoor Tel:(+49)- 0228-287-11649
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Order Form for Molecular Hemostasis

Patient Information (Sticker or written)	Report dispatch address:
Name: _____ **Date of Birth:** _____ **Last Name:** _____ **Gender** ♀ ☐ ♂ ☐ **Adress:**_____	Please provide the complete address for the recipient of the result report.

MATERIAL: 5–10 ml EDTA whole blood
Please label the tube clearly, package securely to prevent breakage, and transport at room temperature.

Clinical suspected diagnosis: (Please include activity/antigen levels)

Familiy history	Additional information	Bleeding	Thrombosis
☐ Yes ☐ No **Degree of relationship:** (please attach anonymized findings if available)	Chromosomal anomaly: Chromosomal sex: ☐ XX ☐ XY	☐ Yes ☐ No If yes: ☐ Spontaneous ☐ Traumatic	☐ Yes ☐ No If yes: ☐ DVT ☐ Other

The samples will be enriched for the entire hemostasis panel. Based on your order, please select:
Complete hemostasis panel / Diagnostic sub-panels / Individual genes.

☐ Complete hemostasis panel or Panel diagnostics (coagulation disorder)

- ☐ Bleeding tendency associated with reduced FVIII activity *(F8, LMAN1, MCFD2, VWF 2N)*
- ☐ Deficiency of Vitamin K–dependent factors *(F2, F7, F9, F10)*
- ☐ Disorders of the fibrinolytic system *(PLAT, PLG, SERPINE1)*
- ☐ Contact phase proteins *(KLKB1, KNG, FXII)*
- ☐ Thrombophilia *(PROC, PROCR, SERPINC1,PROS1)*
- ☐ Thrombomodulin–Protein C system *(PROC, PROCR, PROS1, THBD)*
- ☐ Fibrin-stabilizing factors *(FGA, FGB, FGG, F13A1, F13B)*
- ☐ Hereditary angioedema *(F12 (exon 9), PLG (exon 9), SERPING1)*

Procoagulant Factors (Single factor deficiency)	*Genes*
☐ A-/Hypo-/Dys-fibrinogenemia	*FGA, FGB, FGG*
☐ Faktor II deficiency	*F2*
☐ Faktor V deficiency	*F5*
☐ Faktor VII deficiency	*F7*
☐ Hemophilia A	*F8*
☐ Hemophilia B	*F9*
☐ Faktor X deficiency	*F10*
☐ Faktor XI deficiency	*F11*
☐ Faktor XII deficiency	*F12*
☐ Faktor XIII deficiency	*F13A1, F13B*
☐ Von Willebrand syndrome	*VWF*
☐ Combined FV/FVIII deficiency	*LMAN1, MCFD2*
☐ Congenital ADAMTS13 deficiency	*ADAMTS13*
☐ Kininogen deficiency	*KNG*
☐ Prekallikrein deficiency	*KLKB1*
☐ Combined decificeny of vitamin K dependent factors	*GGCX, VKORC1*

Anticoagulant Factors (Single factor deficiency)	*Genes*
☐ Plasminogen deficiency	*PLG*
☐ Protein C deficiency	*PROC*
☐ Protein C receptor defect	*PROCR*
☐ Protein S deficiency	*PROS1*
☐ Protein Z deficiency	*PROZ*
☐ Antithrombin deficiency	*SERPINC1*
☐ Thrombomodulin defect	*THBD*
☐ PAI-1 defect	*SERPINE1*

Further Diagnostics

- ☐ Thrombophilia polymorphisms
  - ☐ Genetic variants in F5 associated with APCR:
    - ☐ FV-Leiden ☐ FV-Bonn
    - ☐ FV-Hongkong/ FV Cambridge ☐ FV-Nara
  - ☐ Prothrombin G20210A
  - ☐ (Partial) Coumarin resistance/ Partial) Coumarin sensitivity — *VKORC1 Promoter, CYP2C9, CYP4F2 /ggf. F9 Exon 2*

Responsible for the identification of blood samples and the request for laboratory services

_____
Name of the requesting physician (please print in capital letters)

_____ _____
Date **Physician's signature**

Clinic stamp / Referring physician

Tel.:_____ **Fax:**_____

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Order form for molecular genetic analysis of platelet disorders using NGS (Next-Generation Sequencing)

Patient Information (Sticker or written)	**Report dispatch address:**
Name: _____ **Date of Birth:** _____ **Last Name:** _____ **Gender** ♀ □ ♂ □ **Adress:**_____	Please provide the complete address for the recipient of the result report.

The samples will be enriched for the entire hemostasis panel. Based on your order, please select:
Complete hemostasis panel / Diagnostic sub-panels / Individual genes.

The analysis is performed using Next Generation Sequencing (NGS). This technique enables the parallel analysis of numerous genes in a single diagnostic approach, known as panel diagnostics. Our thrombocytopathy/-penia panel consists of 59 genes known to cause the most common inherited forms of thrombocytopenia and thrombocytopathy.

Please check either the complete panel or the desired subpanel below.

☐ **Hemostasis complete panel** **or** **Panel diagnostics (platelet disorder)**

MATERIAL: 5–10 ml EDTA whole blood
Please label the tube clearly, package it securely to prevent breakage, and transport at room temperature.

Platelet morphology			**Platelet Function**	**Clinical Phenotype and Chromosomal Sex**
Platelet Count	**Platelet Size**	**MPV (Mean Platelet Volume)**	**Abnormal aggregation:** ☐ **No** ☐ **Yes** **(if yes, which one)**	
☐ Normal ☐ Decreased ☐ Increased	☐ Normal ☐ Small ☐ Large	☐ Normal ☐ Decreased ☐ Increased	☐ ADP ☐ Collagen ☐ Adrenalin ☐ Ristocetin ☐ Arachidonic acid	Syndromic features:
Clinical suspexted diagnosis: **Family history:** □ **No** □ **Yes** **Degree of relationship:** **(please attach anonymized result letter if available)**				Chromosomal anomaly: Chromosomal sex: □ XX □ XY
Other abnormal laboratory parameters: □ **Flow cytometry:** □ **Immunofluorescence staining:** **(please attach findings if available)** □ **PFA:**				

□ **Platelet Adhesion Disorders**	**Genes**
□ von Willebrand disease, platelet type	*GP1BA*
□ Bernard–Soulier syndrome	*GP9, GP1BA, GP1BB*
□ Leukocyte adhesion defect type III	*FERMT3*
□ **Platelet receptor defect**	**Gene**
□ ADP receptor defect	*P2RY12, P2RY1, P2RX1*
□ GPVI deficiency (collagen receptor)	*GP6*
□ Glycoprotein IV deficiency	*CD36*
□ Thromboxane A2 deficiency	*TBXA2R*
□ Glanzmann thrombasthenia	*ITGA2B , ITGB3*
□ Glycoprotein Ia deficiency	*ITGA2*
□ von Willebrand disease	*VWF*
□ **Platelet secretion disorder**	**Gene**
□ Hermansky–Pudlak syndrome	*HPS1, AP3B1,HPS3-6 DTNBP1, BLOC1S3, BLOC1S6, AP3D1*
□ Bleeding disorder, platelet type 17	*GFI1B*
□ GATA1-associated thrombocytopenia	*GATA1*
□ Chediak–Higashi syndrome	*CHS1, LYST*
□ Gray platelet syndrome	*NBEAL2*
□ Jacobsen/Paris-Trousseau syndrome	*FLI1*
□ Quebec platelet disorder	*PLAU*
□ Griscelli syndrome, type 1/2/3	*MLPH, RAB27A, MYO5A*

Responsible for the identification of blood samples and the request for laboratory services

_____
Name of the requesting physician (please print in capital letters)

_____ _____
Date Physician's signature

□ **Platelet function disorders with cytoskeletal defects**	**Gene**
□ MYH9-associated disorders May–Hegglin anomaly, Sebastian/Fechtner/Epstein syndromes	*MYH9*
□ Wiskott–Aldrich syndrome X-linked thrombocytopenia	*WAS*
□ Filamin A-related disorders with platelet dysfunction	*FLNA*
□ Congenital macrothrombocytopenia	*ACTN1*
□ TUBB1-associated thrombocytopenia	*TUBB1*
□ Scott syndrome	*ANO6*
□ **Andere Thrombozytopathien/penien**	**Gene**
□ Congenital amegakaryocytic thrombocytopenia	*MPL*
□ TCRUS ((thrombocytopenia with radio-ulnar synostosis)	*HOXA11*
□ Autosomal dominant thrombocytopenia 2	*MASTL, ANKRD26*
□ Familial platelet disorder with a predisposition to Acute Myeloid Leukemia.(AML)	*RUNX1*
□ Ghosal syndrome	*TBXAS1*
□ Hereditary hemorrhagic telangiectasia	*ENG, ACVRL1*
□ Tangier disease	*ABCA1*
□ **Further indication-specific diagnostics**	
□ Collagen type 4–associated intracerebral hemorrhages	*COL4A1, COL4A2*

Clinic stamp / Referring physician

Tel.:_____ Fax:_____

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Direktor: Prof. Dr. med. J. Oldenburg
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universitäts klinikum bonn

Consent Form for Genetic Testing

Name, First name (person to be tested):	**Date of birth:**

I consent to the performance of genetic testing in the Hemostasis Panel* and the necessary **blood draw**
☐ from myself ☐ from my child ☐ from the person under my care.
* See the list of genes on pages 1 and 2 of the request form.

I have been **adequately informed** by Dr. med. ----------
about the genetic basis, possibilities of prevention/avoidance/treatment, the purpose, nature, scope, and informative value of the planned investigation as well as the risks associated with blood collection. I was given adequate time for consideration.

Please decide how your sample and the result may be used
(Please check as appropriate. If no selection is made, consent is assumed.)

I would like to be informed about the **results** obtained from the genetic analysis. I have been informed about **my right not to know.**	☐ Yes ☐ No
I would like to be informed about significant **incidental findings** that go beyond the above-mentioned question(s)/condition(s). Only incidental findings with **practical consequences** (e.g., treatment or a concrete preventive program) will be reported.	☐ Yes ☐ No
I would like to be informed about **incidental findings** in genes associated with a **malignant disease** (*RUNX1, ANKRD26, ETV6, GATA1, MPL*).	☐ Yes ☐ No
If the investigations **do not yield a specific result**, I agree that further scientific investigations may be carried out to clarify the coagulation disorder diagnosed in me.	☐ Yes ☐ No
I consent to the storage of **unused test material** for future diagnostic purposes.	☐ Yes ☐ No
I consent to the storage of test material for the p**urpose of verifying results** and **quality assurance.**	☐ Yes ☐ No
I consent to the use of excess test material for the **research** of causes and **treatment improvement** of genetically based diseases. The data may be evaluated in **pseudonymized** form for scientific purposes.	☐ Yes ☐ No
I consent to the storage of test results and records **beyond** the legally required 10-year retention period.	☐ Yes ☐ No
I agree that the results may be used for counseling and testing of **family members**.	☐ Yes ☐ No

This consent can be revoked in whole or in part at any time.

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Place, Date	**Place, Date**
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Signature Patient/Legal representative	**Signature** Responsible physician
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Name (printed)	**Name (printed)**