



Requesting physician:

Patient information:

Request Form

Hypocoagulability / Coagulation defect / Thrombocytopenia / -pathy

Cause of examination: diagnostic predictive for familial mutation _____

Clinical information:

Family history:

(Check all that apply)

- Patient clinically affected
- No previous molecular genetic examinations existent
- The following previous molecular genetic examinations have been done:

Date / Time of sampling:

Signature:

Specimen requirements and logistics:

- Patient Consent Form for Genetic Testing according to the German Law and Request Form
- Billing information (Insurance-, Institutional- or Selfpay-Billing)
- 5 ml EDTA blood collection tube labeled with patient name and date of birth

Patient name: _____ DOB: _____

Hypocoagulability / coagulation defect

- ADAMTS13-deficiency**
ADAMTS13
- Antithrombin III deficiency**
SERPINC1
- Factor II deficiency**
F2
- Factor V deficiency**
F5
- Factor VII deficiency**
F7
- Haemophilia A**
F8
- Haemophilia B**
F9
- Factor X deficiency**
F10
- Factor XI deficiency**
F11
- Factor XII deficiency**
F12
- Factor XIII deficiency**
F13A1, F13B
- Fibrinogen deficiency**
FGA, FGB, FGG
- Hereditary angioedema type 1 & 2**
SERPING1
- Hereditary angioedema type 3**
F12 (Exon 9)
- Flechter factor deficiency**
KLKB1
- Kininogen deficiency**
KNG1
- Hereditary hemorrhagic telangiectasia**
ACVRL1, ENG
- Plasminogen deficiency**
PLG
- Protein C deficiency**
PROC
- Protein S deficiency**
PROS1
- Von Willebrand disease**
VWF

SNP Genotyping

- Antithrombin III Cambridge II A384S**
(rs121909548)
- Prothrombin G19911A**
(rs3136516)

- Prothrombin G20210A**
(rs1799963)
- Factor V Leiden R506Q**
(rs6025)
- Factor V H1299R**
(rs1800595)
- Factor XII C46T**
(rs1801020)
- Factor XIII V34L**
(rs5985)
- β -Fibrinogen G-455A**
(rs1800790)
- FSAP G534E**
(rs7080536)
- MTHFR C677T**
(rs1801133)
- MTHFR A1298C**
(rs1801131)
- PAI-1 A-844G**
(rs2227631)
- PAI-1 4G/5G**
(rs587776796)

Thrombocytopenia/ -pathies

- Bernard Soulier-syndrome**
GP1BA, GP1BB, GP9
- Glanzmann thrombasthenia**
ITGA2B, ITGB3
- Hermansky Pudlak syndrome**
AP3B1, BLOC1S3, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6
- MYH9-related diseases (May-Hegglin-Anomaly (MHA), Sebastian-Syndrome (SBS), Epstein-Syndrome (EPS), Fechtner-Syndrome (FTNS))**
MYH9
- Familial platelet disorder with predisposition to AML**
GATA1
- α - Storage Pool Disease**
NBEAL2, PLAU, VPS33B, VIPAS39, P2RY12
- δ – Storage Pool Disease**
NBEA, AP3B1, BLOC1S3, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6
- $\alpha\delta$ – Storage Pool Disease**
WAS, GATA1
- Thrombocytopenia – large platelets**
GATA1, MYH9, FLI1, NBEAL2, GP1BA, GP1BB, GP9
- Thrombocytopenia – normal platelets**
GATA1, RUNX1, RBM8A
- Thrombocytopenia – small platelets**
WAS
- Wiskott-Aldrich syndrome**
WAS